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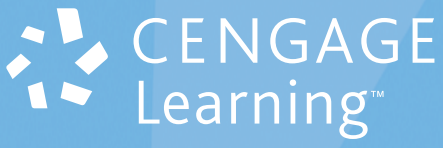
BIOLOGY IN FOCUS

YEAR 12

Glenda Chidrawi
Sarah Bradstock
Elizabeth Thrum
Margaret Robson

2ND EDITION



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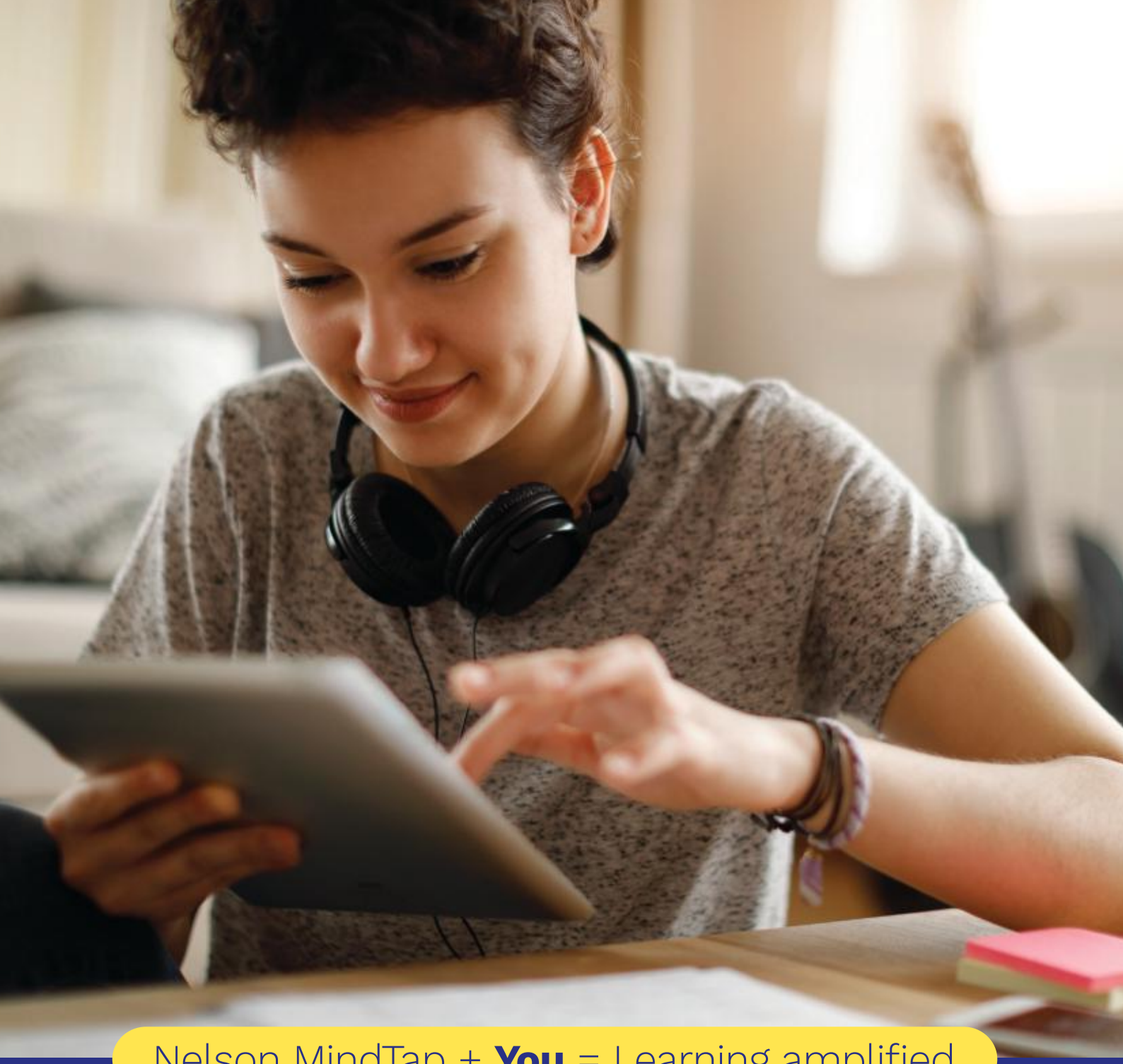

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"I love that everything is interconnected, relevant and that there is a clear learning sequence. I have the tools to create a learning experience that meets the needs of all my students and can easily see how they're progressing."

— Sarah, Secondary School Teacher

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Biology in Focus Year 12
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Contributing author: Sarah Jones
ISBN **9780170485067**

Publisher: Katherine Roan
Project editor: Felicity Clissold
Editor: Kay Waters
Proofreader: Dr Gillian Dite
Indexer: Russell Brooks
Cover designer: Chris Starr (MakeWork)
Text designer: Leigh Ashforth (Watershed Design)
Cover image: Getty Images/Raycat
Permissions researcher: Liz McShane
Content manager: Bradley Smith
Typesetter: MPS Limited

Any URLs contained in this publication were checked for currency during the production process. Note, however, that the publisher cannot vouch for the ongoing currency of URLs.

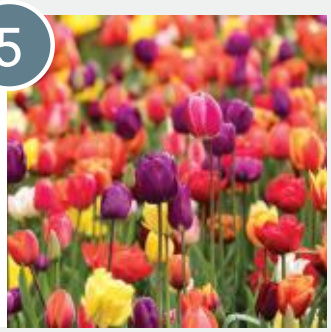
National Library of Australia Cataloguing-in-Publication Data
A catalogue record for this book is available from the National Library of Australia.

Cengage Learning Australia
Level 5, 80 Dorcas Street
Southbank VIC 3006 Australia

For learning solutions, visit **cengage.com.au**

Printed in China by 1010 Printing International Limited.
1 2 3 4 5 6 7 27 26 25 24 23

CONTENTS



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PREFACE

Biology in Focus 2nd edition (Year 12) has been written to the specifications of the NSW Syllabus for the Australian Curriculum, Stage 6 Biology. The authors are academic and classroom teaching experts, chosen for their comprehensive knowledge of the biology discipline and best teaching practice in biology education at secondary and tertiary levels. They have written the text to make it accessible, readable and appealing to students, covering contexts that ensure students gain a wide perspective on the breadth and depth of current biology. The rigorous and methodological approach enables students to reach the highest possible standard. The level of depth and interest are aimed at giving students the necessary understanding to pursue tertiary studies in biology, health sciences, medicine, conservation science, genetics, biotechnology, bioinformatics and other STEM-related courses.

Each chapter of *Biology in Focus* follows a consistent pattern. Learning outcomes from the syllabus appear on the opening page. The text is then broken into manageable sections under headings and sub-headings. All chapters have been structured around the syllabus-identified inquiry questions, and focus within the chapter is on students asking and refining their own questions for investigation. Relevant diagrams that are easy to interpret and illustrate important concepts support the text. New terms are **bolded** and defined in a glossary at the end of the book. Important concepts are summarised to assist students in their note-making. Question sets are found at the end of each section within the chapter, and a comprehensive set of review questions at the end of each chapter expands on the questions sets for further revision and practice. Questions have been set to accommodate the abilities of all students. Complete worked answers appear on the teacher website.

Worked examples, written to connect important ideas and solution strategies, are included throughout the text. Solutions are written in full, including step-by-step instructions on how to perform mathematical calculations. The logic behind each step is explained and mathematical relationships are shown in their biological context. In order to consolidate learning, students are challenged to try similar questions on their own. Answers to these questions appear on the student website.

The broad range of investigations demonstrates the high level of importance the authors attach to exploring and discovering the living world through practical activities and research. Investigations are presented in a manner that provides opportunities for students to develop skills in designing experiments as well as in planning and conducting valid and reliable procedures, taking into account safety and conducting risk assessments. The hands-on activities introduce, reinforce and provide opportunities for students to practise first-hand the Working scientifically investigation skills of the NSW syllabus. Opportunities for students include experimental design, data collection, analysis and drawing conclusions. There are more than enough investigations to meet the 35 hour minimum requirement of the syllabus. Chapter 1 explores the concepts of reliability, validity and the nature of scientific investigation using the scientific method, and provides valuable information for performing and analysing investigations and carrying out depth studies. Detailed information is provided that is designed to enhance students' experiences and to provide them with information that will maximise their marks in this fundamental area, as is reinforced throughout the course.

Students are encouraged to evaluate experimental design and consider ideas for improvement, taking into account accuracy, precision, uncertainty and error – concepts that are introduced in Chapter 1. This invaluable tool supports student learning as they work through chapter questions and investigations.

Biology in Focus 2nd edition (Year 12) provides students with a comprehensive study of modern biology that will fully prepare them for exams and any future studies in the area.

Glenda Chidrawi (lead author)

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AUTHOR AND REVIEWER TEAMS

Glenda Chidrawi – lead author

Glenda Chidrawi (BSc Hons, HDipEd (PG)), with an Honours degree in molecular biology, has been involved in biology education for more than 30 years. She has lectured and taught biology at universities and schools in both South Africa and Australia (Brigidine College 2000–2014 and Knox Grammar School 2015–2016). Glenda is currently an education consultant with the Association of Independent Schools of NSW. Glenda has co-authored senior Biology textbooks, including *Biology Options – Communication*, part of an award-winning series. Glenda has also co-authored the *Biology in Focus Preliminary* and *Biology in Focus HSC* textbooks and the Student Resource CDs and Teachers' Resource CDs that support these publications. Glenda has had articles published in educational journals and has written programs for the AISNSW and material for the NSW QTP Science Program *Teaching the Stage 6 Syllabus*. More recently, Glenda co-authored the *Nelson iScience 10* textbook for the NSW Australian syllabus. Glenda is a familiar figure at education conferences in Australia and currently works in the area of teacher accreditation at AISNSW.

Sarah Bradstock

Sarah grew up in Sydney and graduated from the University of Sydney with a Bachelor of Veterinary Science in 1990. She practised as a small animal veterinarian for eight years. After completing a Graduate Diploma of Education in 1997 she worked as a full-time Science teacher in Sydney for 20 years. She is currently undertaking a Master of Education in Digital Literacy and Knowledge Networks at Charles Sturt University because she has a special interest in the use of digital technology to enhance student engagement and learning in the Biology classroom. Sarah's special biological interest is in animal and human physiology and disease processes.

Margaret Robson

Margaret Robson has been a Science teacher with the NSW Department of Education for the last 44 years, teaching both Biology and Chemistry.

Margaret was one of the co-authors of the very successful *Biology in Focus HSC* textbook. Her work for this textbook also included writing for the Student resource CD and the Teachers' Resource CD. Margaret was a HsC marker for for Biology 25 years, including 9 years as a Senior Marker and 5 years as SOM. She also served as a member of the committee responsible for preparing the Catholic Trial paper in Biology for seven years.

Elizabeth Thrum

Elizabeth has been teaching biology for 27 years in a variety of contexts and schools. She has developed a reputation among her peers as an experienced Biology teacher at the HSC level in both marking and writing of trial papers. She is currently the Head of Science at Knox Grammar School, where she works with a passionate team of educators. She has a passion for teaching Biology and helping students reach their potential. She also likes to work with Biology teachers to ensure that they can teach engaging lessons. Above all she would like students to see how biology is relevant in their daily lives.

ACKNOWLEDGEMENTS

Author acknowledgements

Lead author Glenda would like to acknowledge **Mrs Joyce Austoker-Smith**, who encouraged her to put pen to paper and mentored her through the early years of writing textbooks. The authors would like to thank their families and colleagues for their support and encouragement, their publisher **Eleanor Gregory** for her guidance, and the many students they have taught over the years whose interest in Biology and desire to learn has been their inspiration.

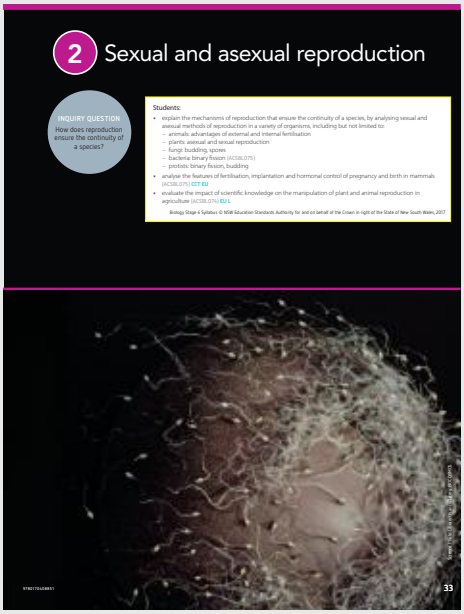
Publisher acknowledgements

Eleanor Gregory sincerely thanks **Glenda**, **Marg**, **Sarah** and **Liz** for their perseverance and dedication in writing this book. She also thanks **Colin Harrison, Deborah de Ridder, Evan Roberts** and **Anita O'Connell** for reviewing the manuscript to ensure that it was of the highest quality. Also thanks to **Gillian Dewar**, **Anita O'Connell**, **Kirsten Prior**, **Marg Robson** and **Rachel Whan** for creating NelsonNet material.

USING *BIOLOGY IN FOCUS*

Biology in Focus Year 12 has been crafted to enable you, the student, to achieve maximum understanding and success in this subject. The text has been authored and reviewed by experienced Biology educators, academics and researchers to ensure up-to-date scientific accuracy for users. Each page has been carefully considered to provide you with all the information you need without appearing cluttered or overwhelming. You will find it easy to navigate through each chapter and see connections between chapters through the use of margin notes. Practical investigations have been integrated within the text so you can see the importance of the interconnectedness between the conceptual and practical aspects of Biology.

The content is organised under four modules, as set out in the NESA Stage 6 Biology syllabus. Each module begins with a **module opening page**.



Each chapter begins with a **chapter opening page**. This presents the learning outcomes from the NESA Stage 6 Biology syllabus that will be covered in the chapter and also gives you the opportunity to monitor your own progress and learning.

To improve comprehension, literacy and understanding, a number of strategies have been applied to the preparation of our text. One of these is the use of shorter sentences and paragraphs. This is coupled with clear and concise explanations and real-world examples. New terms are **bolded** as they are introduced and are consolidated in an end-of-book glossary.

Throughout the text, important ideas, concepts and theories are summarised in **key concept boxes**. This provides repetition and summary for improved assimilation of new ideas.

KEY CONCEPTS
- The genetic consequences of meiosis:
  - One cell undergoes two meiotic divisions to generate four haploid cells.
  - The genes in each haploid cell are a new combination of the parental genes.
  - The new combination results from both crossing over and random segregation, allowing the individual alleles of maternally and paternally derived chromosomes to assort independently.
- The genetic consequences of fertilisation:

Learning across the curriculum content has been identified by NESA as important learning for all students. This content provides you with the opportunity to develop general capabilities beyond the Biology course, and links into areas that are important to Australia and beyond. This content has been identified by a margin icon.

Literacy

Step-by-step instructions on how to carry out particular procedures, such as drawing an annotated diagram or a Punnet square, and the logic behind each step, are provided in **worked examples**. Answers to the problems at the end of the worked example are provided in the back of the book and on the teacher website.

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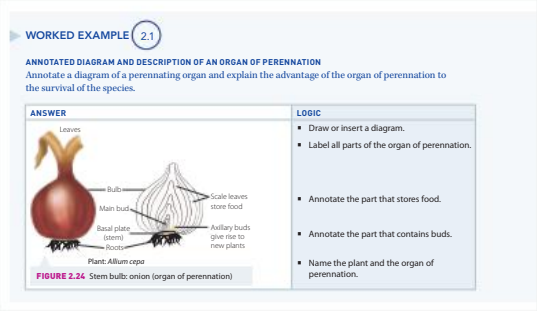


WORKED EXAMPLE 2.1

ANNOTATED DIAGRAM AND DESCRIPTION OF AN ORGAN OF PERENNATION

Annotate a diagram of a perennating organ and explain the advantage of the organ of perennation to the survival of the species.

ANSWER	LOGIC
Leaves, Bulb, Main bud, Basal plate (stem), Roots, Scale leaves store food, Axillary buds give rise to new plants. Plant: *Allium cepa*	▪ Draw or insert a diagram. ▪ Label all parts of the organ of perennation. ▪ Annotate the part that stores food. ▪ Annotate the part that contains buds. ▪ Name the plant and the organ of perennation.

FIGURE 2.24 Stem bulb: onion (organ of perennation)

Biology is a science and you need to be given the opportunity to explore and discover the living world through practical investigations. **Investigations** introduce and reinforce the Working scientifically skills listed in the NESA Stage 6 Biology syllabus. In some cases, the investigations are open ended. These provide you the opportunity to design and carry out your own scientific investigation, either individually or in a group. At times you are prompted to consider ideas for improvement or further investigation to illustrate that science is an ongoing and improving process. At other times, investigations are secondary sourced, meaning that you need to research the subject using data and information gained by other people. Further information on how to conduct a scientific investigation can be found in the Working scientifically and depth study chapter on page 1.

INVESTIGATION 5.1

Information and communication technology capability

Literacy

Weblink Independent assortment and gamete diversity Watch the animation, listen to the narration and then work through the quiz.

Weblink Meiosis with crossing over

Secondary-source and practical investigation to model meiosis, fertilisation and mutations

During meiosis, genetic variation arises as a result of the behaviour of chromosomes during:

- synapsis and crossing over
- independent assortment and random segregation.

In your investigation, you need to model how variation is introduced in the process of meiosis, fertilisation and mutation.

AIMS

1 To model meiosis (including crossing over, independent assortment and random segregation) and predict variations in the gametes produced (Part A)
2 To model fertilisation in order to predict variations in the genotype of offspring (Part B)
3 To predict variations in the genotype of offspring if a mutation was to arise during meiosis and/or subsequent fertilisation (Part C)

RESOURCES

To research how meiosis may be modelled and gain ideas for creative approaches, refer to secondary source material. This may include information in this textbook, as well as online websites, video clips and animations, such as the weblink resources.

PART A: CHROMOSOME BEHAVIOUR DURING MEIOSIS LEADS TO GENETIC VARIATION

AIM

To model meiosis, including crossing over, segregation, independent assortment of chromosomes and the production of haploid gametes

MATERIALS

Pipe cleaners (or playdough/plasticine/strips of paper) in two different colours, to represent chromosomes – one colour represents paternal and the other maternal chromosomes. Make each homologous pair of chromosomes a different length – long, medium and short. To keep your model simple, demonstrate meiosis in a parent cell with *three* pairs of chromosomes.

METHOD

1 In your model of meiosis, use a cell with:
  – at least three pairs of chromosomes made out of pipe cleaners, strips of paper or strings of beads
  – two colours to distinguish between maternal and paternal chromosomes
  – different-sized chromosomes to distinguish between the three homologous pairs (for example: long, medium and short).
2 Move the chromosomes through each stage of meiosis on templates of cells drawn on A3 paper, or use the Modelling meiosis template worksheet. Select a method of recording your results from ideas suggested in the results section.

Model how variation is introduced by the process of meiosis, showing the processes described below.

Meiosis I

1 *Crossing over* – linked genes are exchanged between paternal and maternal chromosomes, increasing the possible combinations of genotypes.

156 MODULE FIVE » HEREDITY 9780170408851

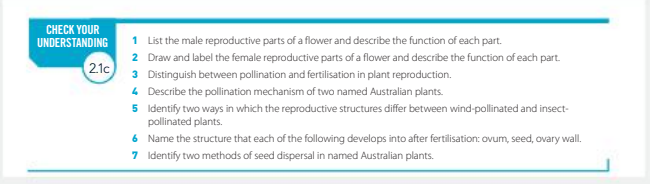
The **risk assessment** table occurs within the investigation. The table highlights the risks of the investigation and provides suggestions on how to minimise these risks – they are not to be considered comprehensive. Teachers are expected to amend this table in the case of substitutions or in the case of any additional risks. This may mean obtaining relevant Safety Data Sheets (SDS) for certain chemicals. All teachers are required to follow the safety guidelines of their specific school and associated government legislation when students are in their care.

RISK ASSESSMENT

Full understanding of a concept is often constructed from many pieces of information. Due to the sequential nature of a book, this information cannot always be presented together because it is best placed in other chapters. Links between concepts that occur on other pages or in other chapters are indicated using the **margin notes**.

You will learn more about fluid transport in Chapter 6.

Regular opportunities to recall new terms and review recent concepts are provided as short **check your understanding** question sets throughout each chapter.

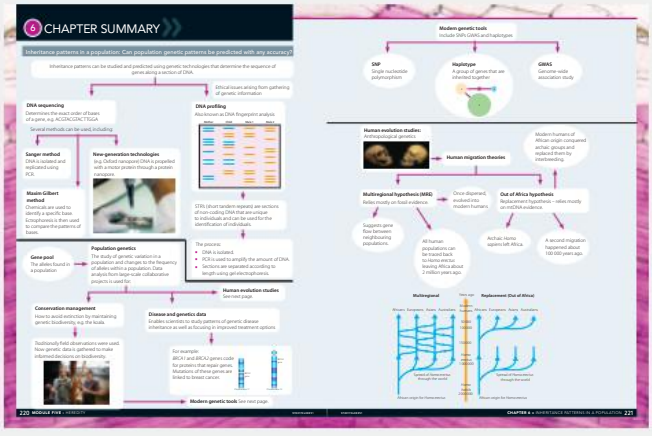


CHECK YOUR UNDERSTANDING 2.1c

1 List the male reproductive parts of a flower and describe the function of each part.
2 Draw and label the female reproductive parts of a flower and describe the function of each part.
3 Distinguish between pollination and fertilisation in plant reproduction.
4 Describe the pollination mechanism of two named Australian plants.
5 Identify two ways in which the reproductive structures differ between wind-pollinated and insect-pollinated plants.
6 Name the structure that each of the following develops into after fertilisation: ovum, seed, ovary wall.
7 Identify two methods of seed dispersal in named Australian plants.

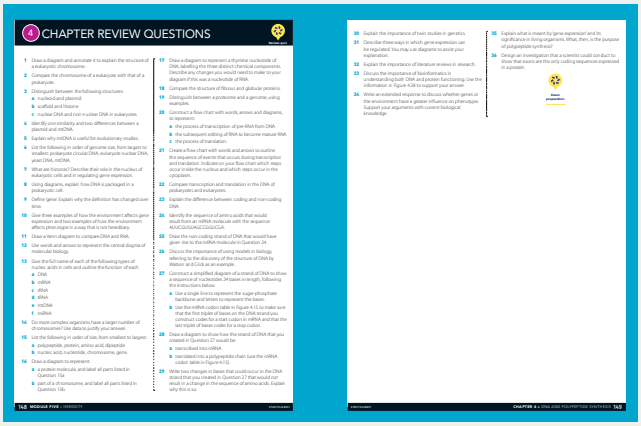
The **chapter review** section, which appears at the end of each chapter, provides:

- a visual **chapter summary** that shows how the important concepts are linked. This will be a valuable tool when you are revising for tests and exams

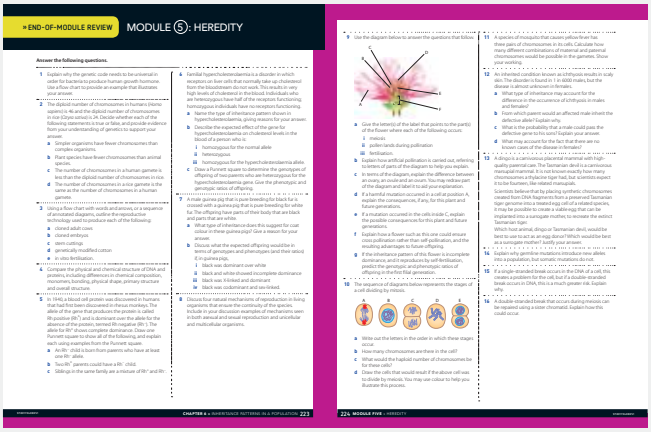


CHAPTER SUMMARY

- **end-of-chapter review questions** that review understanding and provide opportunities for application and analysis of concepts and how they interrelate.



Each module concludes with a **module review**. This contains short-answer questions that provide you with the opportunity to assimilate content across the chapters that fall within that module.



The **depth study** provides you with the opportunity to pursue a topic of interest from within the course. It enables you to study a topic in more depth and present your findings in a format of your choice. Advice and support to assist you in undertaking your depth study can be found in Chapter 1, as well as suggestions for topics provided at the end of each module review. Refer to the NESA Stage 6 Biology syllabus for the full details on the scope and completion of your depth study.

DEPTH STUDY SUGGESTIONS

- Investigate the structure and physiology of a variety of flowering plants, with an emphasis on reproductive mechanisms that are adaptations that ensure continuity of the species.
- Research the claim that sexual reproduction helps speed up evolution and may allow some algae to adapt quickly enough to tolerate the rise in sea surface temperature. Evaluate the possibility that this adaptation will allow some corals to survive a bleaching event.
- Investigate gametogenesis and compare and model differences in meiosis during the production of egg cells and sperm cells in mammals.
- Look into the Red Queen hypothesis and find out whether it promotes natural selection for or against sexual reproduction.

SYLLABUS MAPPING

Working scientifically mapping

Content statements from the NESA Stage 6 Biology syllabus are shown in full on the chapter opening pages of the chapters where they are dealt with. A full mapping of chapters and content statements can be found on the NelsonNet Teacher website. Below is a mapping of the outcome statements for Working scientifically across all the chapters of *Biology in Focus Year 12*.

OUTCOME STATEMENTS	CHAPTER																	
A STUDENT:	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18
BIO11/12-1 develops and evaluates questions and hypotheses for scientific investigation	✓	✓		✓	✓	✓	✓		✓	✓	✓	✓				✓		✓
BIO11/12-2 designs and evaluates investigations in order to obtain primary and secondary data and information	✓	✓	✓	✓	✓		✓			✓	✓	✓				✓		✓
BIO11/12-3 conducts investigations to collect valid and reliable primary and secondary data and information	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
BIO11/12-4 selects and processes appropriate qualitative and quantitative data and information using a range of appropriate media	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
BIO11/12-5 analyses and evaluates primary and secondary data and information	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
BIO11/12-6 solves scientific problems using primary and secondary data, critical thinking skills and scientific processes	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
BIO11/12-7 communicates scientific understanding using suitable language and terminology for a specific audience or purpose	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓

1 Working scientifically and depth studies

A student:

- Questioning and predicting
  - BIO11/12-1 develops and evaluates questions and hypotheses for scientific investigation
- Planning investigations
  - BIO11/12-2 designs and evaluates investigations in order to obtain primary and secondary data and information
- Conducting investigations
  - BIO11/12-3 conducts investigations to collect valid and reliable primary and secondary data and information
- Processing data and information
  - BIO11/12-4 selects and processes appropriate qualitative and quantitative data and information using a range of appropriate media
- Analysing data and information
  - BIO11/12-5 analyses and evaluates primary and secondary data and information
- Problem solving
  - BIO11/12-6 solves scientific problems using primary and secondary data, critical thinking skills and scientific processes
- Communicating
  - BIO11/12-7 communicates scientific understanding using suitable language and terminology for a specific audience or purpose

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Worksheets
- Laboratory rules
- How to successfully evaluate and use websites
- Validity, reliability, accuracy and precision
- Writing a bibliography
- A good practical report

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Science is the systematic study, by observation and experiment, of the natural and physical world (Fig. 1.1). Science is characterised by a way of thinking and working, and, most fundamentally, by questioning. The knowledge and understanding that arise from this questioning are not in themselves science. They are the products of science, as is the technology that arises from this knowledge and understanding. Science is **empirical**, which means that when scientists ask questions, they seek to answer them by using evidence, in particular observational and experimental evidence.

FIGURE 1.1 Investigating in biology

Biology as a field of study was named in the 19th century and arose from the studies of medicine and natural history, both of which date back to ancient times. The term 'biology' comes from the Greek words *bios* (life) and *logos* (word or discourse). Biology asks questions about all living things, including plants, animals and micro-organisms. It asks questions about their structure and functioning, how and why they have changed over time and continue to change, about their interactions with each other and the environment, and about biodiversity and the continuity of life – looking at heredity and variation. These fields of interest in biology are grouped into subdivisions such as botany, zoology, microbiology, evolutionary biology, ecology and genetics. Given that all living things are interdependent, biology is a fascinating science!

1.1 The nature of biology

Questions lead to investigations and these in turn lead to scientific theories that are testable and falsifiable. This applies to all sciences, including biology. For a theory to be considered scientific, it must be possible to test it and, most importantly, to test whether it is not true. This is what **falsifiable** means: 'able to be disproved'. This sets science apart from many other disciplines in which there are theories that cannot be tested or disproved. Such theories are not scientific.

Worksheet
Laboratory rules

This is why scientists never talk about proving a theory, but rather about providing evidence to support a theory. When a large enough amount of evidence has been gathered that supports a theory, then that theory is accepted by the scientific community. Examples of theories in biology that have so much evidence supporting them that they are generally accepted are the Cell Theory and the Theory of Evolution by Natural Selection.

However, no matter how much evidence you gather supporting a theory, it takes only one experiment that disagrees to disprove a theory. As Einstein said: 'No amount of experimentation can ever prove me right; a single experiment can prove me wrong'.

There are many examples of theories and hypotheses in biology that were proposed and later rejected or changed when new evidence came to light. For example, the theory of spontaneous generation is now obsolete, and a theory proposed as the one-gene-one-enzyme theory was later changed when it was realised that a single gene may code for a number of different polypeptides. There are also examples of theories that were based on hoaxes, such as that of Piltdown man. Some hypotheses have been rejected because the scientific method used could not be repeated and was

later shown to be invalid (for example, the hypothesis that proposed that autism could be caused by a certain vaccination).

KEY CONCEPTS

- Scientific theories are falsifiable; they can be disproved, but they cannot be proved. For a theory to be accepted it must be supported by a great deal of evidence.
- A good hypothesis is falsifiable and it takes only one instance of results that disagree with the hypothesis to disprove it.
- No amount of success in testing a hypothesis can prove it is right. Each confirming instance only increases one's confidence in one's idea.

The scientific method – an overview

Critical and creative thinking

The **scientific method** is the process of systematically gathering information and data by observation and measurement, and using the information and data to formulate and test hypotheses. It is from such investigations that the body of scientific knowledge that we accept today has been accumulated.

The scientific method is summarised in Figure 1.2.

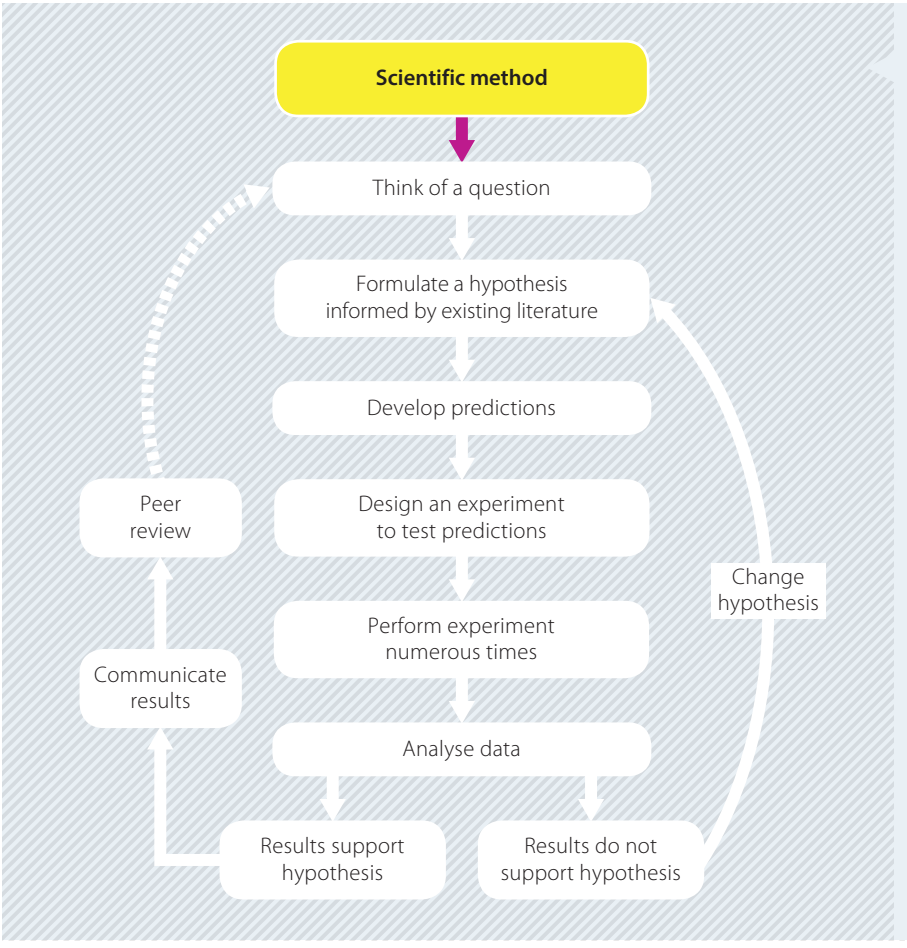


FIGURE 1.2 The scientific method

Hypotheses

The scientific method begins with asking questions (sometimes called research questions). Based on these questions, you formulate a **hypothesis**, which is a tentative answer to your question. This usually

How to formulate a hypothesis is discussed in more detail on pages 11–12.

involves reading the literature to see if anyone has already answered your question or investigated a similar question. For example, we might hypothesise that if we use a particular fertiliser on a certain species of seedling, then the seedlings will grow taller. We could test this hypothesis by performing experiments where we measure the height of a particular species of seedling subjected to different fertilisers.

In scientific investigations, progress is often not a straight line, from one point to the next, but a series of progressions that sometimes veer off the original path. Often, the result of your initial experiments will make you reassess the direction you intend taking, and may lead you to change your hypothesis and refine your experimental design (Fig. 1.2).

Experiment design and validity of results

An experiment is designed and performed to test a prediction, and the results are then analysed. If the results of the experiment agree with the prediction, then the hypothesis is supported. Note that it is not proved, only supported. There may be other explanations that would also be supported by the results. If the results do not agree with the prediction, then the hypothesis is not supported and the scientist needs to come up with another explanation.

Validity, reliability and accuracy are discussed in more detail on page 16.

Experiments are considered **valid** when scientists test the hypothesis that they intended to test and get consistent and accurate results when repeated. A valid experiment involves setting up controls and making sure that the only thing that changes in the experiment is the variable being tested. All other conditions must be **controlled** to remain the same. Experiments are considered **reliable** when they can be repeated to give the same results and random error is eliminated or minimised. An experiment is considered **accurate** when its measurements are close to the true value – for this to be achieved, the risk of error in measurement must be kept to a minimum. For an experiment to be valid, it must be both reliable and accurate. This will be dealt with in more detail in a later section (page 16).

Communication and peer review

Reproducibility and peer review are important aspects of science. If an experiment cannot be repeated to give the same results, then there is a good chance that a mistake was made and the experiment is not valid. For example, if experiments cannot be repeated to give the same results because of uncertainties in the original measurements, the result is that the hypothesis is clearly disproved.

Scientists communicate their work to each other to share new ideas and information and as a way of contributing to the ongoing development of science. They usually communicate new findings to each other in seminars and conferences, as well as writing articles for scientific journals. When you conduct an experiment and write a report on it, the report is very much like a scientific paper.

Before a scientific paper is published, it is reviewed by other scientists – experts in the particular area – who evaluate it. They try to determine whether:

- the experiments conducted were appropriate
- the conclusions drawn were valid
- the hypothesis is clearly supported or not.

If the paper is considered to make a useful contribution to science, and the experiments and analysis are valid, then it will be published. Other scientists can then read the paper and use it to inform their own work. Scientists also communicate their work in other ways to the public and to students.

Descriptions of the scientific method are somewhat idealised. In practice, the scientific method may be a bit messy and not follow the steps in order. Sometimes scientists have only questions but no hypothesis to answer them. In these cases, experiments are done or observations are made to try to form a hypothesis that can then be tested. Sometimes, in trying to answer one question, a new and more interesting question arises, so a scientist will change the experiments to work on the new question instead. However, once a scientific discovery is made, and even when a new and exciting discovery is made by accident, the scientific method will be used to formulate and test hypotheses that arise to explain it.

Weblink
Science and pseudoscience
Read this article about the scientific method and come up with your own explanation of the difference between science and pseudoscience.

KEY CONCEPTS

- A hypothesis is a predictive statement about the relationship between the variables in an investigation and is an 'expected' answer to a question.
- The scientific method consists of questioning and formulating hypotheses, making measurements to test the hypotheses, analysing the results, and communicating them for peer review. It is the process by which science proceeds.

Biology as a scientific discipline

Disciplines within science can be characterised by the sorts of questions that they ask. Biology asks questions about the organisation and grouping of living organisms, how living things change over time, why some species survive and others do not, and how living things interact with their environments and with each other.

Biologists find that these questions may be answered by looking at the morphology and functioning of living organisms, how they reproduce and are adapted for survival, their origin, distribution and interactions, as well as inheritance patterns and increases in diversity. Biological studies ask questions at various levels – macroscopic, microscopic and molecular. Developments in technology and advances in knowledge and understanding in biology progress hand in hand, with each one assisting the other.

The more we find out in biology, the more questions are generated. There are many questions we have not answered yet. As current and future biologists investigate and answer these questions, yet more questions will arise that no one has thought of yet. Will all the possible questions about living organisms ever be answered?

Models in science

Over the years, scientists have asked questions and sought and, at times, found answers to those questions. From their answers we have constructed **models** of how living things may be related and how and why they change over time. These models are always changing, as we get more evidence and better answers to existing questions, or as we seek answers to new questions. Models are representations of biological reality – they are not the reality itself any more than a model aeroplane is a real aeroplane. Models can be physical models, some are mathematical models made up of equations and data, and yet others are conceptual models consisting of principles, laws and theories. Biologists use all sorts of models as they ask and seek to answer questions. At times they combine models and switch between models.

Models in biology have two important purposes – to explain how things work, and to predict what will happen. A model that does not accurately predict the results of an experiment will generally be revised or replaced. Two models may give similar results in some situations but different results in others. Model selection is important to get valid and reliable results. For example, the model of Mendelian (autosomal recessive) inheritance and the model of non-Mendelian inheritance can both be used to describe and analyse the pattern of inheritance of genetic traits. Overall, the two models give similar results, with exceptional circumstances taken into account in non-Mendelian genetics. For example, in the Mendelian genetics model, all genes are assumed to be inherited independently of each other and are either dominant or recessive. However, it has been found that some genes occur on the same chromosome or are located on sex chromosomes, and breeding experiments that involve these genes do not give the expected ratios typical of the Mendelian model. In studies of other genes, no clear dominance is seen – both genes are expressed if present, or a blend of genes is expressed. In these cases, non-Mendelian models such as sex-linkage, co-dominance or incomplete dominance are used to analyse patterns of inheritance, taking the additional complexities into account. This doesn't mean that either model is always 'right' or 'true', just that Mendelian inheritance is the basic model, but the other inheritance models may need to be applied to take into account further complexities in inheritance patterns. Choosing the right model for a situation is an important skill in solving problems in biology.

See Chapter 5 for more details on these models of inheritance.

KEY CONCEPTS

- Biology uses models such as physical, mathematical and conceptual models to describe biological systems and to make and test predictions. Models are constantly being refined as we learn more.

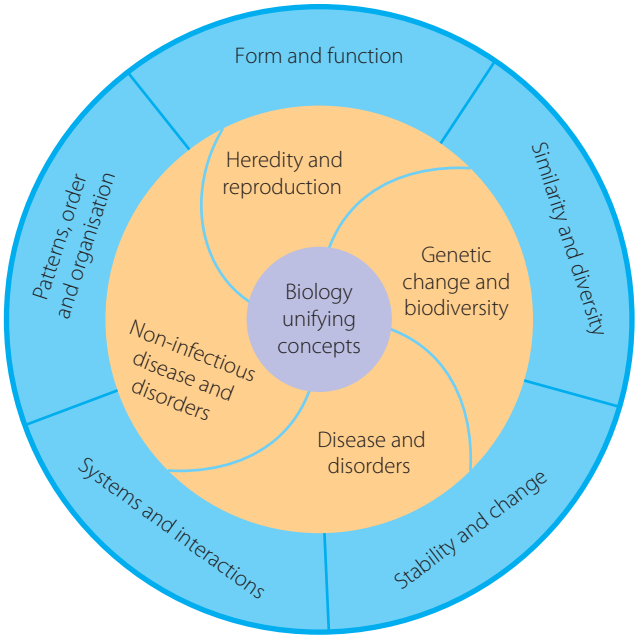
Biology – knowledge and understanding

As you progress through your Biology course, you will learn a lot of useful skills, and practise working scientifically by performing investigations and depth studies. You will also gain some knowledge and develop a deeper understanding of biology.

The knowledge that has arisen from answering questions asked by biologists can be broadly categorised into five major concepts: form and function; similarity and diversity; stability and change; systems and interactions; and patterns, order and organisation. Figure 1.3 shows how the five concepts in the Year 12 Biology course fit into unifying themes.

As you learn more of the content knowledge of biology, you need to create your own mental models to help you understand it. Concept maps are a useful way of representing your mental models. They help to remind you that biology is not simply a collection of facts. Every idea in biology is connected to other ideas. For each module in biology, you should create a concept map both to record the content that you learn and to make connections between different content areas and modules.

FIGURE 1.3 Unifying concepts in biology



1.2 Solving scientific problems: depth studies

Depth studies are your opportunity to work scientifically and solve scientific problems. When performing a depth study, you will pose questions, develop hypotheses to answer your questions, and then seek evidence to support or disprove your hypotheses. The evidence may come from the existing scientific literature or from your own experiments. You will need to analyse data to determine whether your hypotheses are supported. Analysing data usually requires you to represent it in some way, often mathematically or graphically. Finally, as scientists do, you need to communicate your findings to others.

There are many ways that you can do this, and you need to choose the method most appropriate to the audience you wish to communicate with.

Types of depth studies

There are two broad types of depth study:

1. first-hand or practical investigations, where you design and perform experiments to gather primary data, or to test a claim or device
2. investigations based on secondary sources, where you research and review information and data collected by other people.

First-hand investigations to gather primary data may be:

- work undertaken in a laboratory
- field work, where observations are undertaken at home, school or elsewhere (for example, on excursions or by engaging with community experts)
- the creation and testing of a model or device.

Secondary-source depth studies may include:

- undertaking a **literature review**
- investigating emerging technologies and their applications in biology
- analysing a science fiction movie or novel
- developing an evidence-based argument or a historical or theoretical account.

Depth studies may be presented in different forms, some of which include:

- written texts (experiment reports, field work reports, media reports, journal articles, essays and management plans)
- visual presentations (diagrams, flow charts, keys, posters and portfolios)
- multimedia presentations
- physical models
- a combination of the above.

All depth studies will involve the analysis of data, either from **primary data** that you collect yourself or **secondary data** that you collect from analysing other people's research such as longitudinal data or resource management data. Looking for patterns and trends in data will involve analysing and constructing graphs, tables, flow charts, diagrams, keys, spreadsheets and/or databases. This will be dealt with in more detail in the section Designing your investigation (see page 15). You may also wish to refer to the NSW Stage 6 Biology syllabus document for more information.

How to write a literature review is discussed in more detail on pages 9–10.

Why undertake a depth study?

Depth studies encourage us to identify areas of interest and enable us to deepen our understanding in a chosen area, taking responsibility for our own learning. Although a field of study may be identified by the teacher, students may pursue their own area of interest within this field, be it technology, current research, biologists working in the field, or other areas.

Depth studies provide students with time and an opportunity to:

- use the research methods that scientists use
- analyse works for scientific relevance and validity
- broaden their range of reading in a field of interest
- extend their depth of thinking and understanding
- ask questions and investigate areas that do not have definite answers
- investigate contentious issues and use critical thinking skills to consider the validity of views expressed in a variety of sources

How to plan a depth study is discussed in detail on pages 12–15.

- use inquiry-based learning and develop their creative thinking in an area of their own choosing, at their own level.

Stages in a depth study

The summary below outlines the four main stages of conducting a depth study, and the *Working scientifically* skills (see the NESA Stage 6 Biology syllabus) that you will need to develop and apply at each stage.

1 *Initiating and planning* involves:

Questioning and predicting

(BIO-1): develops and evaluates questions and hypotheses for scientific investigation

Planning investigations

(BIO-2): designs and evaluates investigations in order to obtain primary and secondary data and information

2 *Implementation and recording* involves:

Conducting investigations

(BIO-3): conducts investigations to collect valid and reliable primary and secondary data and information

Processing data and information

(BIO-4): selects and processes appropriate qualitative and quantitative data and information using a range of appropriate media

3 *Analysing and interpreting* involves:

Analysing data and information

(BIO-5): analyses and evaluates primary and secondary data and information

Problem solving

(BIO-6): solves scientific problems using primary and secondary data, critical thinking skills and scientific processes

4 *Communicating* involves:

(BIO-7): communicates scientific understanding using suitable language and terminology for a specific audience or purpose

Working Scientifically Skills Outcome: BI011/12-1

Posing questions and formulating hypotheses


Jim West / Alamy Stock Photo

FIGURE 1.4 Brainstorm as many ideas as you can in your group.

The first step to beginning any investigation or depth study is deciding on a question. A good research question is one that can be answered by conducting an experiment, making observations or conducting a secondary-source investigation.

Obviously, it is a good idea to investigate something that you find interesting. If you are working in a group, try to find something that is interesting to everyone in the group.

A good way to start is by 'brainstorming' for ideas (Fig. 1.4). This applies whether you are working on your own or in a group. Write down as many ideas as you can think of. Avoid being critical at this stage. Get everyone in the group to contribute, and accept all contributions uncritically. Write every idea down.

After you have run out of ideas, it is time to start being critical. Decide which questions or ideas are the most interesting. Think about which of these it is actually possible to investigate given the time and resources available. Remember that the most important resources you have are the skills of the people in the group. Make a shortlist of questions, but keep the long list too, for the moment. Once you have your shortlist, it is time to start refining your ideas.

A good **research question** should define the investigation, set boundaries and provide some direction to the investigation. The difference between developing a research question and formulating a hypothesis can be summed up as 'known versus unknown'. You need to do some research of known results in your area of interest (research questions) before deciding what you think the expected outcome of an experiment may be (hypothesis).

Personal and social capability

Critical and creative thinking

Weblink
Six methods of data collection and analysis

Writing a literature review – refining your question

Your depth study will be from one of the areas described in Figure 1.3, which is based on the NSW Stage 6 Biology syllabus document. These areas are described in the remaining chapters. However, you will need to go beyond the basic syllabus content because the purpose of a depth study is to extend your knowledge while building your skills at working scientifically.

The next step is therefore to find out what is already known about the ideas on your list. You need to do a literature review. If your depth study is a secondary-source investigation, then the literature review may be the investigation itself. A formal written literature review includes the information you have found and complete references to the sources of information. It also includes interpretation and critique of what you have read. This is particularly important for a secondary investigation.

Literacy

Information and communication technology capability

Critical and creative thinking

Why are literature reviews important?

Literature reviews are important because they help you to:

- increase your breadth of knowledge and identify what is and is not known about an area of research
- learn from others and think of new ideas that may be relevant to a research project
- identify gaps in current knowledge that you may wish to research or recommend be researched by scientists in the future
- identify methods that could be relevant to your project (avoid reinventing the wheel and/or making the same mistakes as others)
- identify the variety of views (sometimes opposing views) in an area of research and consider how these fit in with your own views.

Your literature review

A literature review is a search and evaluation of available literature in a particular subject area. It has a particular focus and is always defined by your research question or hypothesis.

The process of conducting a literature review involves researching, analysing and evaluating the literature. It is not merely a descriptive list of the information gathered on a topic, or a summary of one piece of literature after another. It outlines any opposing points of view in the research and also expresses the writer's perspective of the strengths and weaknesses of the research being reviewed. A literature review brings together results of different studies, pointing out areas where researchers or studies agree, where they disagree, and where significant questions remain. By identifying gaps in research, literature reviews often indicate the direction for future research.

When planning an investigation, you will find that using a literature review will give you an idea of past findings, procedures, techniques and research designs that have already been used. This will help you to decide which methods are worth copying, which need modifying and which to avoid (those that have been inconclusive or invalid). You may plan your investigation to target a gap in research or try to replicate an investigation to test and validate claims and ideas.

The length of your literature review will depend on its purpose. If it is a depth study in itself, it will need to be more detailed and draw conclusions about the research. If it is used as an introduction to inform your research, it will be shorter and more focused. Discuss this with your teacher.

Reasons to write a literature review

- To extract information from sources
- To consider the validity of views expressed in each source
- To consider how existing views fit in with a research project, to place it in context and demonstrate how the research is linked to a body of scientific knowledge
- As an end in itself (for example, as a secondary-source research assignment to use the findings to support a concluding judgement)
- As a starting point to begin planning a primary investigation, identifying both what is known and where there are current gaps in research

Characteristics of a good literature review

- Helps the reader know what knowledge and ideas have been established on a topic and the areas of strength and weakness in the research
- Organises the information gathered into sections that present themes
- Does not attempt to list all published material, but rather brings together and evaluates the literature according to a question, hypothesis or guiding concept

How to write a literature review

1. Getting started: define the topic or research questions (key concepts) and formulate a literature review question (you may have to do some wide reading before finalising this step). Write a list of key words.
2. Find articles: use library catalogues, databases and the Internet. Refine your search technique, using specific words that narrow your search to the focus question. Interpret and evaluate your search results. Record search words that are successful and, if necessary, modify your search strategy.
3. Structure and write your literature review:
   - i Introduction: define the topic, establish your reasons for reviewing the literature, state the specific focus of the review and explain the organisation or sequence of your review.
   - ii Body: group the literature according to common themes, provide an explanation of the relationship between the research question and the literature reviewed, and proceed from the general, wider view of the research to the specific area you are targeting.

     Include information about the usefulness, recency and major authors or sources of the literature.
   - iii Conclusion: summarise important contributions of the literature, point out important flaws or gaps in research if appropriate, and explain the link between your focus question and the literature reviewed (if the literature review is your depth study) or why you have chosen your area of investigation (if the literature review was conducted to refine your investigation).

Worksheet
How to successfully evaluate and use websites

Weblink
Literature reviews
More detail on how to write a good literature review

Weblink
The CRAAP test
Apply the CRAAP test to any websites that you find

Evaluating sources

Critical and creative thinking

Always be critical of what you read. Be wary of pseudoscience, and any material that has not been peer reviewed. Apply the CRAAP (currency, relevance, authority, accuracy, purpose) test to websites that you find. The most reliable sites are from educational institutions, particularly universities, and government

and scientific organisations such as the CSIRO and WHO, and professional journals such as the *Medical Journal of Australia* and international equivalents. You can narrow your search to particular types of sites by including in your search terms 'site:edu' or 'site:gov' so that you find sites only from educational or government sources.

Make sure you keep a record of the information that you find as well as the sources, so that you can correctly reference them later (Fig. 1.5). It is a good idea to start a logbook (page 18) at this stage. You can write in references or attach printouts to your logbook. This can save you a lot of time later on. Your logbook may be hard copy or electronic but, either way, begin keeping it now.

Finally, talk to your teacher about your ideas. They will be able to tell you whether your ideas are likely to be possible for investigation given the equipment available. They may have had students with similar ideas in the past and can make suggestions about what worked well and what did not.

After you have researched your questions and ideas, you will ideally be able to narrow the shortlist down to the one question that you want to tackle. If none of the questions or ideas looks possible (or interesting), then you need to go back to the long list.

FIGURE 1.5 Start researching your topic and make sure you keep a record of all your references. Good record keeping is important in scientific research, and it begins at this stage of the investigation.

Proposing a research question or hypothesis

If you are doing a primary-source investigation (of, for example, the effect of the environment on phenotype), then you need to define a research question and/or hypothesis.

For example, you may begin by thinking: 'I wonder if a new fertiliser will affect plant growth'. This idea is too general, so you need to turn it into a more specific research question. The research question may be: 'What effect does a new fertiliser have on root and stem growth in a plant?' The question needs to be specific enough to guide the design of your experiment. It needs to include what you will be varying (for example, type of fertiliser) and what result you will expect (root and/or stem growth). Once you have decided on your research question, further reading will guide you to design a suitable experiment (Fig. 1.6). You would read up about the chemical components of different fertilisers, ways of measuring root and stem growth, and what types of plants have and have not had growth benefits from fertilisers in the past. You need to decide on what specifically you will be changing (for example, you may decide to select a new fertiliser with twice as much nitrogen as the old fertiliser) and exactly what you will measure (shoot height, root length and root mass in a seedling). The research question can then be turned into a hypothesis: *If a fertiliser that contains more nitrogen is introduced, then a particular seedling's shoot height, root length and root mass will increase.*

FIGURE 1.6 You need to frame your research question carefully. This student is investigating plant growth with different fertilisers.

KEY CONCEPTS

- Frame your research question carefully by making it specific enough to guide the design of the investigation.
- Poor research question: 'How can we make a seedling grow the best?' 'Best' is a vague term. What you mean by 'best' may not be what someone else means.
- Good research question: 'Which one of two fertilisers gives the maximum growth of roots and stem in a seedling?' This question is not vague. It tells you what you will be varying and what you will be measuring. It also gives a criterion for judging whether you have answered the question.

Formulating a hypothesis

A hypothesis is a predictive statement about the relationship between the variables and is an 'expected' answer to your question. It is often written as an 'If … then …' statement, to explain an expected relationship, such as: 'If *x* is introduced/increased/decreased, then *y* will increase/decrease/stay the same.'

An example of a hypothesis is:

If the amount of nitrogen in the fertiliser provided to a seedling in the soil is increased, *then* the height of the stem and/or length of the roots of the seedling will increase.

Your hypothesis should give a prediction that you can test, ideally quantitatively (that is, by taking measurements).

A hypothesis is usually based on an existing model or theory. It is a prediction of what will happen in a specific situation based on that model. For example, investigators may use a model that describes how, in the nitrogen cycle, nitrates from the soil are assimilated into proteins for plant growth. A hypothesis based on an increased nitrate model may predict that, with plants that normally grow in nitrogen-poor soils, introducing additional nitrogen in a fertiliser may increase growth.

A good research question or hypothesis identifies the variables that will be investigated. Usually you will have one dependent variable and one independent variable. For a depth study you may have two or more independent variables that you control (for example, you may test two different fertilisers if time and resources allow). You should only change one variable at a time in any investigation.

If your experiments agree with predictions based on your hypothesis, then you can claim that they support your hypothesis. This increases your confidence in your model, but it does not prove that it is true. Hence, an aim for an experiment should never start 'To prove …', because it is not possible to actually prove a hypothesis, only to disprove it.

If your experimental results disagree with your hypothesis, then you may have disproved it. This is not a bad thing! Often the most interesting discoveries in science start when a hypothesis based on an existing model is disproved, because this raises more questions.

Even if your question or hypothesis meets these criteria, do not be surprised if you change or modify it during the course of your investigation or depth study. In scientific research, the question you set out to answer is often only a starting point for more questions.

KEY CONCEPTS

- Investigations begin with a question, which is used to formulate a hypothesis.
- A literature review helps you refine your question or hypothesis. It helps you know what knowledge and ideas have been established on a topic and the areas of strength and weakness in the research.
- A good hypothesis is a statement that predicts the results of an experiment, states the expected relationship between the variables and can be tested using quantitative measurements.

1.3 Planning your depth study

Critical and creative thinking

There are many things to consider when planning and designing an investigation. You need to think about how much time you will have, what space and equipment you will need, and where you will go if you want to make measurements or observations outside. If you are doing a secondary-source investigation or some other type of depth study, such as a creative work (e.g. building a physical model), you still need to plan ahead to make sure you have the resources you need.

You may be working in a group or on your own. Most scientists work in groups. If you can choose who you work with, think about it carefully. It is not always best to work with friends. Think about working with people who have skills that are different from your own.

Work and enterprise

Having a plan allows you to ensure that you collect the data, whether from a primary or secondary source, that is needed to test your hypothesis. The longer the investigation, the more important it is that you have a clear plan. Table 1.1 lists several things to consider.

TABLE 1.1 Planning your depth study

PRIMARY-SOURCE INVESTIGATION	SECONDARY-SOURCE INVESTIGATION
What data will you need to collect?	What information will you need to gather?
What materials and equipment will you need?	What sources will you use?
When and where will you collect the data?	When and where will you gather the information?
If you are working in a group, what tasks are assigned to which people?	If you are working in a group, what tasks are assigned to which people?
Who will collect the data?	Who will collect what information?
Who will be responsible for record keeping?	How will record keeping be done to avoid plagiarism?
How will the data be analysed?	How will the information be analysed?
How will sources be referenced?	How will sources be referenced?

Devising a plan for your investigation

Working Scientifically Skills Outcome: BIO11/12-2

Personal and social capability

The most common problem that students have is time management. It is important to plan to have enough time to perform the experiments, including repeat measurements, and to analyse them and report on them.

A good plan will help you keep on track. Your teacher may ask you to hand in a plan of your depth study before you begin the implementation stage. Table 1.2 gives an idea of the types of things you should think about.

TABLE 1.2 Depth study plan

INTRODUCTION TO DEPTH STUDY PLAN	
Title *What?*	Choose a title for your depth study.
Rationale *Why?*	Explain why you have chosen this area of research. Describe what you are hoping to achieve through this investigation. Include any ways you think your investigation may benefit yourself, your class and possibly your family, friends and the school/wider community (if applicable).
Type of depth study and research model (if applicable) *Which?*	State the type of depth study you intend conducting (e.g. literature review, practical investigation). Where applicable, describe any theoretical models that you will use for your depth study. Include references to your reading and explain why you chose this model.
Timeline	
Action and time frame *When?*	Working scientifically skills How?
1 Initiating and planning *When?* (For example, weeks 1–2)	*Questioning and predicting*: formulate questions and/or a hypothesis; make predictions about ideas, issues or problems. *Planning*: wide reading – research background information; assess risks and ethical issues; plan valid, reliable and accurate methods; select appropriate materials and technologies; identify variables; plan experimental controls and how to measure them.
2 Implementation and recording *When?* (For example, weeks 2–4)	*Conducting investigations*: safely carry out valid investigations; make observations and/or accurate measurements; use appropriate technology and measuring instruments. *Processing and recording data and information*: collect, organise, record and process information and/or data as you go.
3 Analysing and interpreting *When?* (For example, week 4–mid week 5)	*Analysing data and information*: reduce large amounts of data by summarising or coding it; begin looking for trends, patterns or mathematical relationships. *Problem-solving*: evaluate the adequacy of data (relevance, accuracy, validity and reliability) from primary and/or secondary sources.

INTRODUCTION TO DEPTH STUDY PLAN	
4 Communicating *When?* (For example, weeks 5–mid week 6).	*Presenting your depth study*: use appropriate language, scientific terminology, calculations, diagrams, graphing and other models of representation; acknowledge your sources.
Final presentation	Due date: end of week 6
Data collection	
Note that what you submit in your final depth study may be different from your initial planning list.	
a **Action: independent variable** Describe what you will change in your investigation.	**b** **Outcome: dependent variable** What will you measure and how will you measure it? (Quantitative/ qualitative data?)
c **Validity: controlled variables** What will you need to keep constant to make this a fair test? What control(s) will you use (if applicable)?	
Data analysis and problem solving	
d **Data analysis** What method(s) will you use to analyse the data, and how will you represent the trends and patterns?	**e** **Conclusion** How will you judge whether the experiment was valid? How will your data allow you to test your hypothesis or answer your question?

Working Scientifically Skills Outcome: BIO11/12-3

Selecting equipment

A well-framed question or hypothesis will help you choose the equipment that you need. For example, if your hypothesis predicts a temperature change of 0.5°C, then you will need a thermometer that can measure to at least this precision (precision and accuracy are discussed on page 19). You also need to know how to use the equipment correctly. Always ask if you are unsure. The user manual will usually specify the precision of the device and let you know of any potential safety risks, so read it.

Work and enterprise

Personal and social capability

You need to think about how you can minimise uncertainties and errors. Minimising uncertainty is not just about using the most precise equipment you can find; it is also about clever experimental technique.

Working safely: risk assessment

You may be required to complete a risk assessment before you begin your investigation. You need to think about three things.

1. What are the possible risks to you, to other people, and to the environment or property?
2. How likely is it that there will be an injury or damage?
3. If there is an injury or damage to property or environment, how serious are the consequences likely to be?

A 'risk matrix' can be used to assess the severity of a risk associated with an investigation. 'Negligible' may be getting clothes dirty. 'Marginal' might be a bruise from falling off a bike, or from a broken branch in a tree. 'Severe' could be a more substantial injury or a broken window. 'Catastrophic' would be a death or the release of a toxin into the environment. You need to ensure that your investigation is low risk.

Once you have considered what the possible risks are, you need to think about what you will do about them. What will you do to minimise them, and what will you do to deal with the consequences if something does happen? You can use a risk assessment table like the one shown in Table 1.3.

Consider where you will perform your experiments or observations. Will you need to consider the convenience or safety of others? Talk to your teacher about what space is available.

TABLE 1.3 Example risk assessment table for managing risks

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THE RISK?
Potassium permanganate	Eye irritant	Wear safety glasses. If the solution comes in contact with the eyes, use an eyewash.
Glassware	Broken glass may cut the skin	Handle all glassware with care. If glass breaks, sweep it up with a brush and dustpan.

RISK ASSESSMENT

Ethical considerations

There are ethical frameworks for biological investigations, protecting the lives of animals and humans. You need to take into consideration ethical principles before you begin your research. Think about basic human values, animal rights, the rights of children, and whether the use of some technologies has ethical repercussions. Ethical principles in biology could be a whole depth study of its own.

In your research for your literature review, include information about ethical codes of conduct related to your investigation.

In a secondary-source investigation, take precautions with cyber safety and remember to keep your personal information private.

Ethical understanding

Weblink
Use of live organisms in schools

KEY CONCEPTS

- In primary-source investigations, you collect and analyse your own data. In secondary-source investigations, you analyse someone else's data.
- Investigations need to be planned carefully so that they answer your research question. You also need to consider safety, ethical issues and the possible environmental impacts of your investigation.

Designing your investigation

Working Scientifically Skills Outcome: BIO11/12-3

Data: reliability, accuracy, validity and relevance

When designing your investigation, think about how you can minimise uncertainties and overcome failure. For example, root growth will be affected if plant roots are attacked by fungus (mould). Try to think of all the things that could go wrong in your experiment and put preventative measures in place. You may also need to come up with a backup plan, so start early in case things go wrong and you need to re-do your experiment.

If you are conducting a secondary-source investigation, then your literature review will be the basis of your investigation. Remember that a literature review is not simply a summary of what you have read – you need to add meaning. This may come from comparing and contrasting competing models and constructing an argument, or by analysing and presenting secondary-source data. When using secondary sources, remember to make comparisons between data and claims in a number of reputable sources, including science texts, scientific journals and reputable Internet sites, and to reference these appropriately.

If you are doing a primary-source investigation, a brief literature review will form the background information and you will then make measurements to gather data yourself. You can collect data by performing experiments or making observations in the field. You will gain practice at making measurements if you do some of the investigations in the following chapters. These investigations can form the basis of your depth study.

Information and communication technology capability

Critical and creative thinking

Literacy

Variables

When doing experiments, you need to decide which variable you will change, what you will measure and which variables you will control. Consider which variables you can control, and which you cannot. Typically, an experiment will have three types of variable:

1. an **independent variable**, which we are testing and we therefore purposefully change
2. a **dependent variable**, which is the result that we measure – this changes as a result of changing the independent variable. We assume that the dependent variable is in some way dependent on the independent variable
3. **controlled variables**, which are kept constant so that they do not interfere with our results.

Reliability

Whenever possible you should make repeat measurements. This allows you to check that your measurements are reliable. Your results are reliable if repeat observations and/or measurements taken under exactly the same circumstances give the same results within experimental uncertainty. If a result is not **reproducible**, it is not a reliable result. The cause may be that a variable other than the one you are controlling is affecting the value of the dependent variable. If this is the case, you need to determine what this other variable is, and control it if possible. Results may also be unreliable if random errors occur in the method. A reliable experiment is one which, if repeated multiple times, gives the same result (within an acceptable margin of error). Reliable sources, such as scientific journals and texts, are sources whose information is trustworthy because they are is written by qualified professionals and are consistent across multiple sources.

Accuracy

Accuracy may refer to a result or to an experimental procedure. Accuracy of a result (data) is a measure of how close it is to an expected value given in scientific literature (for example, scientific journals). Secondary-source information is accurate when it is found to be similar to information presented in peer-reviewed scientific journals.

To improve accuracy in experiments, we use the most precise measuring instruments available, avoid human error (for example, measuring errors), carry out repeat trials, and find an average to smooth out random errors so that the value we obtain approaches the expected value more closely.

Accuracy is also linked to any uncertainty in measurement. For example, we can determine the size of red blood cells by estimating their number in a field of view and dividing by the size of that field of view. Alternatively, we can measure their size with less uncertainty using a mini grid slide or a calibrated digital microscope.

Plausible accuracy is accuracy that is estimated, taking into consideration the evident sources of error and the limitations of the instruments used in making the measurements.

Validity

To ensure that results are valid, in a primary investigation you must carry out a fair test.

- Identify variables that need to be kept constant.
- Develop and use strategies to ensure these variables are kept constant.
- Demonstrate the use of a control.
- Use appropriate data collection techniques.
- Trial procedures and repeat them, checking that the results are the same each time.

In a control, you remove the factor being tested in the experiment to see whether, without that factor, a different result is obtained.

These steps ensure that the process used and the resultant data measure what was intended. Results need to be valid if you are going to be able to draw a conclusion from them.

An investigation is valid if factors that may vary within an experiment are deliberately held constant to ensure a fair test. These 'controlled variables' are kept the same so that the only factor that is allowed to change in the experiment is the independent variable. The result is that the investigation measures what was intended and the data obtained is accurate and reliable.

Evaluating your investigation

Some good questions to ask to assess reliability, validity and accuracy are shown in Table 1.4.

TABLE 1.4 Assessing reliability, accuracy and validity in investigations

	PRIMARY INFORMATION AND DATA	SECONDARY INFORMATION AND DATA
Reliability	Have I tested with repetition and obtained consistent results? Have I done multiple trials and found an average to eliminate random errors?	How consistent is the information with information from other reputable sources? Are the data presented based on repeatable processes?
Accuracy	Do the results of the investigation agree with the scientifically accepted value? Have I used the best measuring equipment available?	Is this information similar to information presented in peer-reviewed scientific journals?
Validity	Does my experiment measure the variable of interest? Does it actually test the hypothesis that I want it to? Have all variables apart from those being tested been kept constant? Have errors been kept to a minimum? Are my results accurate and reliable?	Do the findings relate to the hypothesis or problem? Are the findings accurate and the sources reliable?

KEY CONCEPTS

- An experiment will have three types of variables: dependent, independent and controlled.
- Reliability of first-hand data is the degree to which repeated observation and/or measurements taken under identical circumstances yield the same results.
  - To *assess* reliability, compare results from repeat experiments to see if they are the same.
  - To *improve* reliability, control all variables other than those being tested, repeat and average results to reduce random errors, and use precise measuring equipment so that the same result can be obtained each time the experiment is repeated.
- To *assess* accuracy, examine how close a measurement is to its true value *or* how similar the information is to that in peer-reviewed scientific literature.
  - To *improve* accuracy, minimise uncertainty, reduce systematic errors and use the most precise measuring equipment available. Use peer-reviewed secondary sources.
- To *assess* validity in a primary investigation, evaluate how closely the processes and resultant data measure what was intended.
  - In a secondary investigation, assess whether the information is relevant to the topic and if it is from reliable sources.
  - To *improve* validity, refine the experiment design to reduce complex variables that cannot be kept constant, as well as reducing random and systematic errors.

Gathering data

Numeracy

You also need to consider how many data points to collect. In general, it is better to have more data than less. However, you will have limited time to collect your data, and you need to allow time for analysis and communicating your results. A minimum of 6–10 data points is usually required to establish a relationship between variables, if the relationship is linear. A linear relationship is one where if you plot one variable against the other you get a straight line. If you think the relationship might not be linear, then take more data points and think carefully about how they will be spaced. You should try to collect more data in the range where you expect the dependent variable to be changing more quickly. For example, if you are measuring temperature of a hot object as it cools as a function of time, then you should collect more data early, when cooling is more rapid.

You will need to keep a record of what you do during your investigation. Do this in a hard copy or electronic logbook.

Keeping a logbook

Literacy

Scientists keep a logbook for each project that they work on. A **logbook** is a legal document for a working scientist. If the work is called into question, then the logbook acts as important evidence. Logbooks are sometimes even provided as evidence in court cases (for example, in patent disputes). Every entry in a scientist's logbook is dated, records are kept in indelible form (pen, not pencil), and entries may even be signed. Never record data on bits of scrap paper instead of your logbook!

Your logbook will include:

Personal and social capability

- notes taken during the planning of your investigation
- a record of when, where and how you carried out each experiment
- diagrams showing the experimental set-ups, biological drawings, and other relevant information
- all your raw results
- all your derived results, analysis and graphs
- all the ideas you had while planning and carrying out experiments, and analysing data
- printouts, file names and locations of any data not recorded directly in the logbook.

It is not a neat record, but it is a complete record (Fig. 1.7). Your teacher may check your logbook at various intervals to assess your progress.

FIGURE 1.7 Make sure you keep an accurate record of what you do as you do it.

Your logbook

Always write down what you do as you do it. It is easy to forget what you did if you do not write it down immediately. Your logbook may be hard copy or electronic. Either way, your logbook is a detailed record of what you did and what you found out during your investigation. Make an entry in the logbook every time you work on your depth study.

Logbooks are important working documents for scientists. All your data should be recorded in a logbook, along with all records of your investigations.

Recording data and creating scientific tables

Literacy

If you are going to be collecting multiple data points, then it is a good idea to draw a table to record them in. Scientific tables are always drawn with a ruler, and they are fully enclosed tables with appropriate headings. Label the columns in the table with the name and units of the variables. If you know that the uncertainty in all your measurements is the same, then you can record this at the top of the column as well. Otherwise, each data entry should have its uncertainty recorded in the cell with it. When constructing a results table, put units in the headings and not in the body of the table. It is best practice to put the independent variable in the first column and the dependent variable in the second, if you are drawing a vertical table. For a horizontal table, the independent variable is placed in the top horizontal row and the dependent variable in the first vertical column.

Working Scientifically Skills Outcome: BIO11/12-4

It is a good idea to start your analysis while you are collecting your data. If you spot an **outlier** and you are still making measurements, then you have the opportunity to repeat that measurement. If you made a mistake, then put a line through the mistake and write in the new data.

Plotting and analysing data as you go is sometimes beneficial because it allows you to spot something that may be of interest early on in your investigation. You then have a choice between revising your hypothesis or question to follow this new discovery, or continuing with your plan. Many investigations start with one question and end up answering a completely different one. These are often the most fun, because they involve something new and exciting. Some of the most significant finds (for example, penicillin) have come from unexpected results of experiments or serendipity.

Accuracy, precision and errors in measurement

Working Scientifically Skills Outcome: BIO11/12-5

When making measurements, your aim is to be as precise and accurate as possible.

An accurately measured result is one that represents the 'true value' of the measured quantity as closely as possible. When we take repeated measurements, we assume that the mean (average) of the measurements will be close to the 'true value' of the variable. However, this may not always be the case. For example, if you have ever been a passenger in a car with an analogue speedometer and tried to read it, your reading will be consistently different from what the driver reads. This is because of parallax error. The needle sits above the scale, and when viewed from the side does not line up correctly with the true speed. Beware of parallax error with any equipment using a needle. This is an example of a **systematic error**, in which measurements differ from the true value by a consistent amount. Note that often we do not know what the 'true value' is.

Scientists should be aware of the possibility of error in all stages of an investigation. Notes on possible sources of error should be kept in the logbook.

- Planning stage: errors may arise as a result of limitations of time and/or materials. Assess the possibility and adjust the method so that errors are minimised.
- Data collection and processing stages: remember to assess the degree of uncertainty and to keep note of the accuracy of measuring devices.
- Concluding stage: evaluate the validity of the investigation and discuss any sources of error, as well as possible ways of reducing error in future investigations.

Critical and creative thinking

Worksheet
Validity, reliability, accuracy and precision

Sometimes it is difficult to remember the difference between accuracy and precision. For example, on a dart board, think of accuracy as how close to the centre your dart hits, and your measurement of precision as how closely you can group your shots (Fig. 1.8).

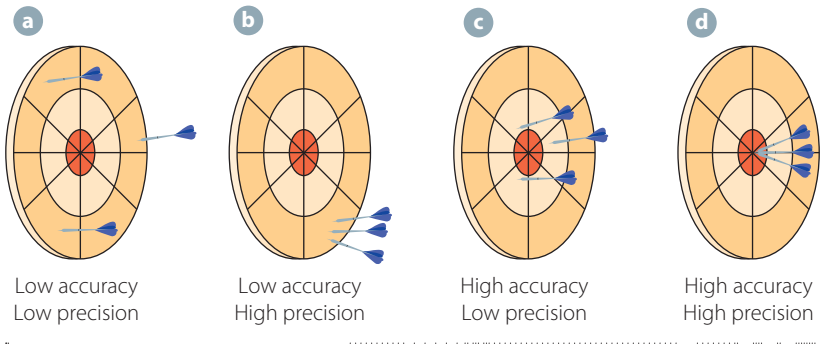


FIGURE 1.8 On a dart board, accuracy is determined by how close to the centre (bullseye) your dart lands. Precision is how closely you can group your darts.

When looking at precision and accuracy in scientific measurements, measurements that are close to the known value are said to be accurate, whereas measurements that are close to each other are said to be **precise**. Therefore, for measurements to be accurate *and* precise, they must be close to the mean value and the measurements need to be close to each other.

A graph may also be used to show the relationship between accuracy and precision (Fig. 1.9).

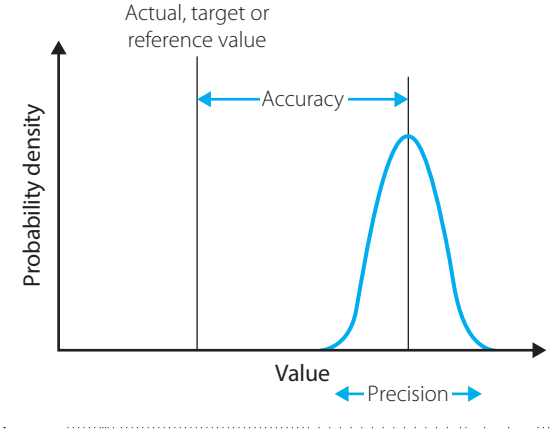


FIGURE 1.9 Graph distinguishing between accuracy and precision

Numeracy

In the scientific definition, precision is described as a measure of the variability of the measurements, so it affects the spread of the repeated measurements about the mean value. The smaller the spread, the greater the precision. This is shown in Figure 1.10. Figure 1.10a shows precise measurements, and Figure 1.10b shows less precise measurements. Note that both data sets are centred about the same mean, so they have the same accuracy.

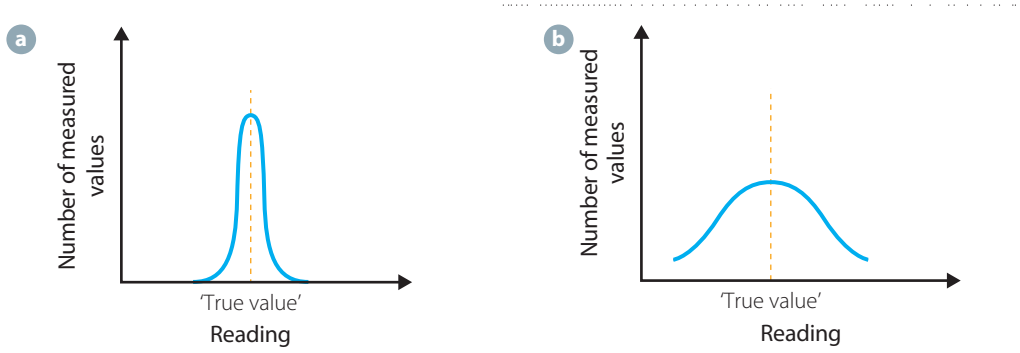


FIGURE 1.10 In a plot of number of measured values versus reading, the results in **a** have a small spread about the mean (true value) and are therefore more precise. The results in **b** have a larger spread about the mean and are less precise.

There is always the risk that errors in measurement may arise when actually doing the measuring, but some errors also arise when we are calculating derived data. We need to keep both types of error to a minimum if our results are to be reliable, accurate and valid.

Weblink
Accuracy and precision

Visit the weblink *Accuracy and precision* to increase your understanding of minimising error and to clarify some concepts about processing of raw data that may seem complex at first. This weblink deals with precision and accuracy, and gives an easy but realistic example of how and why it is necessary to process raw data and to calculate percentage, mean and standard deviation.

Critical and creative thinking

Estimating uncertainties

When you perform experiments, there are typically several sources of uncertainty in your data. Sources of **uncertainty** that you need to consider are the:

- limit of reading of measuring devices
- precision of measuring devices
- variation of the **measurand** (the variable being measured).

For all devices there is an uncertainty due to the limit of reading of the device. The limit of reading is different for analogue and digital devices.

People often confuse precision with the resolution of a measuring device. The resolution tells us about the 'degree to which an instrument can be read'. Precision is the 'degree to which an instrument can be read repeatably and reliably'.

Analogue devices have continuous scales and include swinging needle multimeters and liquid-in-glass thermometers. For an analogue device, the **limit of reading**, sometimes called the resolution, is half the smallest division on the scale. We take it as half the smallest division because you will generally be able to see which division mark the indicator (needle, fluid level, etc.) is closest to. So, for a liquid-in-glass thermometer with a scale marked in degrees Celsius (Fig. 1.11a), the limit of reading is 0.5°C.

Digital devices such as digital multimeters and digital thermometers have a scale that gives you a number. A digital device has a limit of reading uncertainty of a whole division. So a digital thermometer that reads to whole degrees (Fig. 1.11b) has an uncertainty of 1°C. For a digital device the limit of reading is always a whole division, not a half, because you do not know whether it rounds up or down, or at what point it rounds. The digital device (Fig. 1.11c) has a greater resolution than (a) or (b) because it measures to one decimal place. The limit of reading is 0.1°C.

Weblink
Resolution and precision

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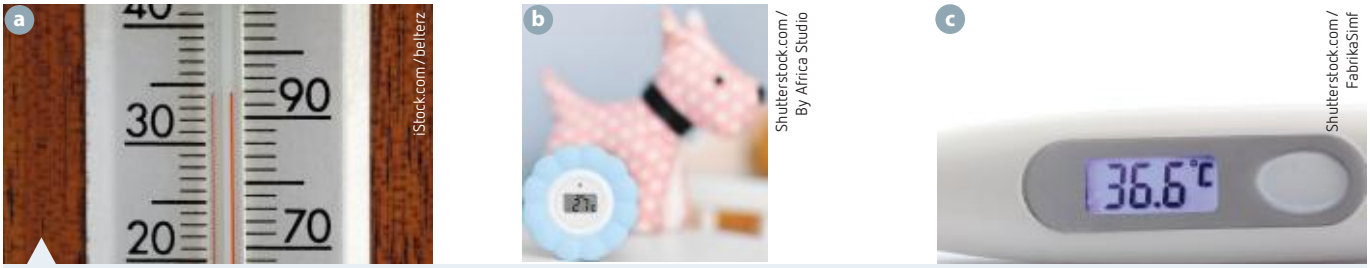

FIGURE 1.11 **a** Analogue and **b** and **c** digital thermometers with different resolutions (reading limits)

The resolution, or limit of reading, is the minimum uncertainty in any measurement. Usually the uncertainty is greater than this minimum.

Measuring devices such as data loggers usually have their precision given in the user manual.

Many students think that digital devices are more precise than analogue devices. This is often not the case. A digital device may be easier for you to read, but this does not mean it is more precise. The uncertainty due to the limited precision of the device is generally greater than the limit of reading (see Figs 1.11a, 1.11b).

KEY CONCEPTS

- Systematic error is due to the measuring device (for example, if it is not calibrated correctly) or technique (for example, parallax error or through incorrect positioning of the instrument).
- Random error is due to unavoidable variations in reading a measurement.
- Accuracy refers to the closeness of a measured value to a standard or known value.
- Precision refers to the closeness of two or more measurements to each other.
- The uncertainty in any measurement depends upon the limit of reading of the measuring device, and the precision of the device.

Analysing data

Working Scientifically Skills Outcome: BIO11/12-5

When you have collected all your data, you will need to analyse it. Record all your analyses in your logbook. If you have more than a few data points, it is a good idea to display them in a table. Tables of data need to have headings with units for each column, and a caption stating what the data means or how it was collected (see how to construct a table on page 18). Tables are used for recording raw data and also for organising derived data.

Critical and creative thinking

Calculating derived data from raw data

Numeracy

Raw data is what you actually measured (with units and uncertainties). Derived data is data that you have calculated using raw data. For example, your raw data may be the length of roots and height of shoots in each plant. From this data you may choose to derive the average length and height of roots and shoots and/or you may wish to calculate the overall percentage growth.

Drawing and using graphs

If you look at any science journal, you will see that almost every article contains graphs. Graphs are not only a useful way of representing data, but they are also commonly used to analyse relationships between variables. You should have lots of graphs in your logbook as part of your exploration of the data. It is often useful to plot your data in different ways, especially if you are unsure what relationship to expect between your dependent and independent variables.

Graphs should be large and clear. The axes should be labelled with the names of the variables and their units. The independent variable is placed on the *x*-axis and the dependent variable on the *y*-axis. Choose a scale so that your data takes up most of the plot area. This will often mean that the origin is not shown in your graph. Usually there is no reason why it should be. The scale is plotted in equal increments.

Weblink
Data points
Some helpful advice on deciding the number of data points

To determine a relationship you need to have enough data points and the range of your data points should be as large as possible. A minimum of six data points is generally considered adequate if the relationship is expected to be linear (give a straight line), but always collect as many as you reasonably can, given the available time.

For non-linear relationships, you need more data points.

Types of graphs

There are different types of graphs so you need to know which type to use. Your choice will depend on what you have measured.

Scatter plots are used when you are looking for a relationship between variables. A scatter plot is a graph showing your data as points (Fig. 1.12a). Do not join them up as in a dot-to-dot picture. Use a line of best fit if they appear to fall in a straight line.

Line graphs are also used to find a relationship between variables. When both the independent and dependent variables are continuous, a line graph is drawn to show how one variable will affect the other. For example, the independent variable may be the number of hours seedlings were exposed to light and the dependent variable may be the average height of the shoots (see Table 1.5).

TABLE 1.5 Height of shoots in seedlings exposed to variable periods of light

TIME EXPOSED TO LIGHT PER DAY FOR 2 WEEKS (hours)	HEIGHT OF SHOOTS (mm)
0	24
2	18
4	17
6	15
8	13
10	11
12	9
14	7

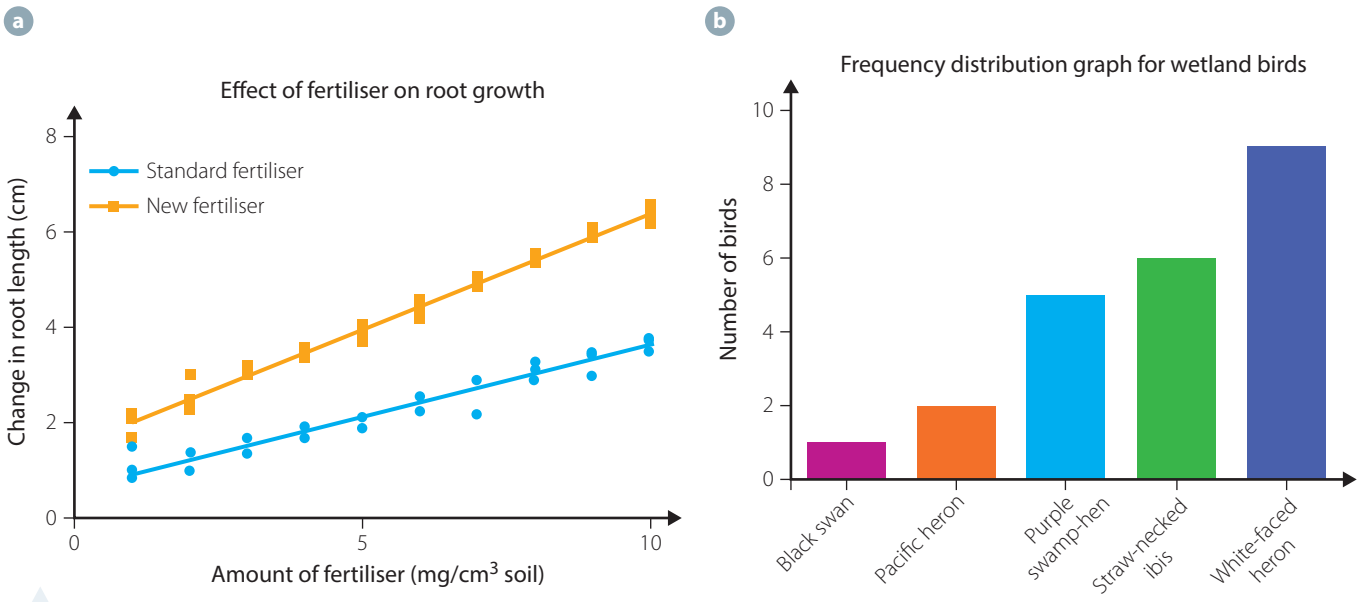

FIGURE 1.12 **a** A scatter plot demonstrating a mathematical relationship; **b** a column graph displaying results from counting categories

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Column or bar graphs are used if groups of things have been counted or measured. An example is the heart rates of different types of animals. A bar graph has categories on the *y*-axis and numbers on the *x*-axis. A column graph has numbers on the *y*-axis and categories on the *x*-axis (Fig. 1.12b). The columns or bars have a gap between them and do not touch.

A **histogram** is similar to a column graph, but the columns touch each other. These are used for data where a continuous variable has been divided into consecutive categories. An example is the average monthly rainfall in Sydney during autumn and winter in 2016 (Table 1.6).

TABLE 1.6 Average rainfall in Sydney during autumn and winter, 2016

MONTH	AVERAGE RAINFALL (mm)
March	130
April	129
May	120
June	133
July	97
August	81

A **sector or pie graph** is used to compare parts of a whole. An example is the composition of a predator's diet (Table 1.7 and Fig. 1.13). A protractor must be used when drawing a pie chart.

TABLE 1.7 Components of diet of red fox (*Vulpes vulpes*)

FOOD	DIETARY CONTRIBUTION (%)
Rabbits	30
Lambs	20
Small marsupials	10
Frogs	10
Lizards	10
Birds	5
Carrion	5
Fruit	5
Other	5

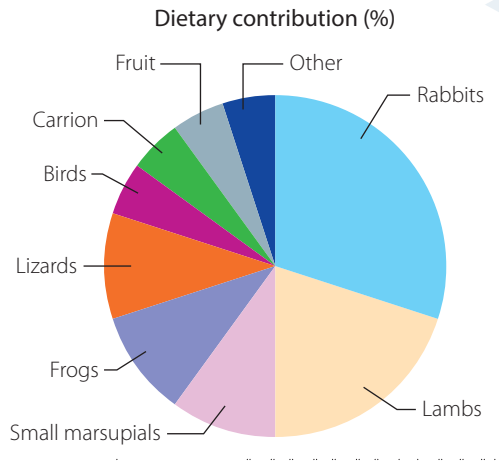

FIGURE 1.13 Data in Table 1.7 shown as a pie graph

Linear lines of best fit

A good graph to start with is simply a scatter plot of the raw data. You will usually be able to tell by looking whether the graph is linear. If it is, then fit a straight line (**line of best fit**).

Removing outliers

Numeracy

When you plot your raw data, you may find that one or two points are outliers. These are points that do not fit the pattern of the rest of the data. These points may be mistakes. For example, they may have been incorrectly recorded or a mistake may have been made during measurement. They may also be telling you something important. For example, if they occur at extreme values of the independent variable, then it might be that the behaviour of the system is linear in a certain range only. You may choose to remove or ignore outliers when fitting a line to your data, but you should be able to justify doing so.

Non-linear lines of best fit

Relationships between variables are often not linear. If you plot your raw data and it is a curve, then do not draw a straight line through it. In this case you need to think a little harder. If your hypothesis predicts the shape of the curve, then try fitting a theoretical curve to your data. If it fits well, then your hypothesis is supported.

Note that a line of best fit is not the same as joining the dots. It is rarely useful or appropriate to join the dots, even though this is often the default setting in spreadsheet software.

KEY CONCEPTS

- Data is usually recorded in tables.
- Graphs are used to represent and analyse data.
- Linear graphs are useful for analysing data.

Interpreting your results

Working Scientifically Skills Outcome: BIO11/12-6

Numeracy

Once you have analysed your results, you need to interpret them. This means being able to either answer your research question or state whether your results support your hypothesis.

You need to take into account the uncertainties in your results when you decide whether they support your hypothesis. For example, suppose you have hypothesised that the maximum range in which the enzyme pepsin will function is between 35°C and 40°C, yet your results show that the maximum activity occurs at 42°C. You may think that this result does not support your hypothesis. To say whether the result agrees with the prediction, you need to consider the uncertainty. If the uncertainty is 1°, then the results disagree with the hypothesis. If the uncertainty is 2° or more, then the results do agree and the hypothesis is supported.

If your hypothesis is not supported, it is not enough to simply say 'our hypothesis is wrong'. If the hypothesis is wrong, what is wrong with it?

Weblink
Enzyme model
Effect of temperature on catalase activity

It may be that you have used a model that is too simple. For example, when designing an experiment using catalase, you may base your hypothesis on the model that enzymes in the human body work best at around human body temperature (37°C), like many digestive enzymes. In your experiment, you then find that the optimum temperature for catalase is 10°C. This may be because you were not aware of the model that proposes that, in order to function in cells lining the digestive tract, catalase needs to avoid being digested. To do this, it changes shape to its functional form and works best at a low optimum temperature between 0°C and 10°C. At body temperature catalase is indigestible but not the optimum shape for peak performance. Therefore, your hypothesis may be better described by a model that takes into account the idea that catalase must avoid being digested and achieves this by functioning best at temperatures lower than body temperature. Before you decide that the model is at fault, however, it is a good idea to check carefully that you have not made any mistakes.

It is never good enough to conclude that 'the experiment didn't work'. Either a mistake was made or the model used was not appropriate for the situation. It is your job to work out which. In doing so, you will come up with more questions.

Experiments that do not support predictions based on existing models are crucial in the progress of science. It is these experiments that tell us there is more to find out, and inspire our curiosity as scientists.

KEY CONCEPTS

- You must know the uncertainty in your results to be able to test your hypothesis.
- If your hypothesis is disproved, you need to be able to explain why.

1.4 Communicating your understanding

Working Scientifically Skills Outcome: BIO11/12-7

Literacy

If research is not reported on, then no one can learn from it. An investigation is not complete until the results have been communicated. Most commonly, a report is written. Scientists also use other means to communicate their research to each other, such as posters and talks. To communicate their work to the public, scientists may use science shows and demonstrations, public lectures, websites, videos and blogs. All of these are useful ways of communicating your understanding, and you need to select the mode that best suits the content you wish to communicate and the audience to whom you wish to communicate.

Writing reports

A report is a formal and carefully structured account of your investigation or depth study. It is based on the data and analysis in your logbook. However, the report is a summary. It contains only a small fraction of the information that you collected.

A report consists of several distinct sections, each with a particular purpose. When writing a report for a science journal, you will need to provide an abstract and an introduction, but for secondary school purposes the following headings are suggested:

- Background information
- Aim
- Hypothesis
- Risk assessment
- Materials
- Method
- Results and analysis
- Discussion of results
- Conclusion
- Acknowledgements
- References
- Appendix.

Reports in scientific journals are always written in the past tense, because they describe what you have done. They start with an abstract – a very short summary of the entire report, typically between 50 and 200 words. The abstract appears at the start of the report but is always the last thing that you write. Try writing just one sentence to summarise each part of your report.

At school level, your report may be written in the present or past tense. Start with a clearly stated aim, making sure it includes variables and the change you will be measuring.

Background information

The background information tells the reader why you did this investigation or depth study and how you developed your research question or hypothesis. This is the place to explain why this research is interesting. The introduction also includes the literature review, which gives the background information needed to be able to understand the rest of the report. The introduction for a secondary-source report is similar to that for a primary-source investigation. In both cases, it is important to reference all your sources correctly.

Aim

In any experiment, the aim describes what you intend to do. For example, you may aim to investigate, to measure, to model, to compare, to verify or to calculate. The aim should be brief and it should link with the hypothesis that predicts what you expect to find.

Hypothesis

The hypothesis is written as a predictive 'If … then …' statement of your expected result, and it must be falsifiable (that is, it must be able to be disproved). It does not give a reason why you expect that result.

Risk assessment

Risk assessment is discussed on pages 14–15.

Materials

A list of all the materials and equipment (including quantities and concentrations, if applicable) that you used during the experiment is provided.

Method

The method summarises what you did. It says what you measured and how you measured it. It also explains, briefly, why you chose a particular method or technique. The method is written in point form. If in the present tense, each sentence usually starts with a verb. The method also describes how you measured your results and recorded your information.

For a primary-source investigation, the method describes how you carried out your experiments or observations in enough detail that someone with a similar knowledge level could repeat your experiments. It should include large, clear diagrams of equipment set-up, such as water baths in enzyme experiments. You should have diagrams in your logbook, but these are generally rough sketches. Diagrams should be redrawn neatly for a report, as in Figure 1.14.

The method section for a secondary-source investigation is generally shorter. If you are doing a review of the current literature on a topic, then your method will say what literature searches you carried out and how you decided which sources to use.

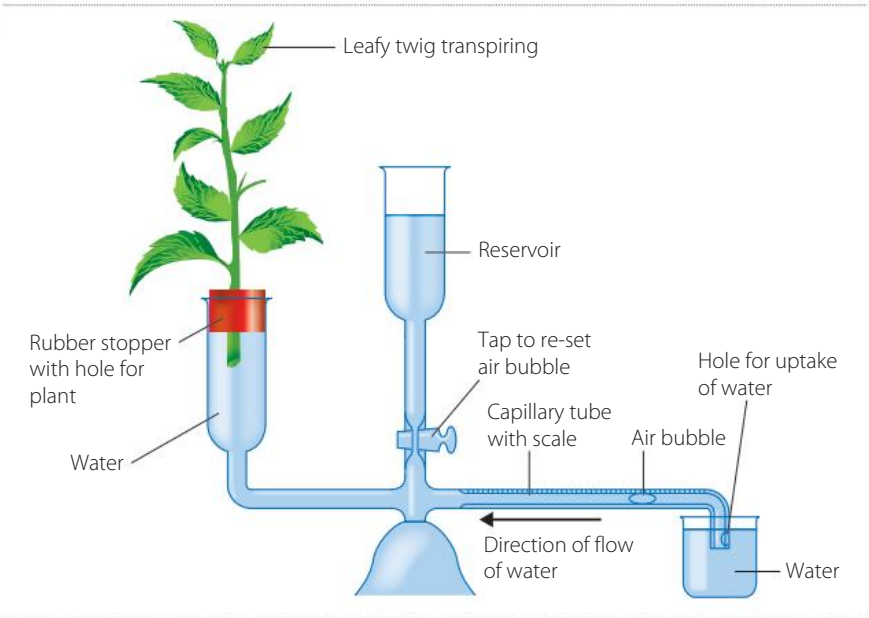

FIGURE 1.14 A potometer is used to measure the rate of absorption in relation to transpiration.

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Results

The results section is a summary of your results. It is usually combined with the analysis section, although they may be kept separate.

Tables comparing the results of different experiments or secondary sources are useful. Avoid including long tables of raw data in your report. Wherever possible, use a graph instead of a table. If you need to include a lot of raw data, then put it in an appendix attached to the end of the report.

Think about what sort of graph is appropriate. If you want to show a relationship between two variables, then use a scatter plot. Display your data as points with uncertainty bars and clearly label any lines you have fitted to the data. Column and bar graphs are used for comparing different data sets. Do not use a column or bar graph to try to show a mathematical relationship between variables.

Any data and derived results should be given in correct SI units with their uncertainties. If you performed calculations, then show the equations you used. You might want to show one example calculation, but do not show more than one if the procedure used is repeated.

Discussion

The discussion should summarise what your results mean. If you began with a research question, give the answer to the question here. If you began with a hypothesis, state whether or not your results supported your hypothesis. If not, explain why. If your investigation led you to more questions, as is often the case, say what further work could be done to answer those questions.

Conclusion

The conclusion is a very brief summary of the results and their implications. Say what you found. A conclusion should be only a few sentences long and should not contain any inferences. Make sure your conclusion relates back to your aim and hypothesis. This is where you state whether or not your hypothesis is supported.

Acknowledgements and references

Scientific reports often include acknowledgements thanking people and organisations that helped with the investigation. This includes people who supplied equipment or funding, as well as people who gave you good ideas or helped with the analysis. In science, as in other aspects of your life, it is always polite to say 'thank you'.

The final section of a report is the reference list. It details the sources of all information that was actually used to write the report. This list will generally be longer for a secondary-source investigation. Wherever a piece of information or quotation is used in your report it must be referenced at that point. This is typically done either by placing a number in brackets at the point (for example, [2]), or the author and year of publication (for example, (Smith, 2016)). The reference list is then provided in either a footnote at the end of each page or a single complete list at the end of the report. There are different formats for referencing, so check with your teacher about what format they prefer. There are several good online guides to referencing.

References versus bibliographies

A reference list is not the same as a bibliography. A bibliography is a list of sources that are useful to understanding the research. The sources may or may not have been used in the report. You should have a bibliography in your logbook from the planning stage of your investigation. The references will be a subset of these sources. A primary-source investigation does not include a bibliography. A secondary-source investigation may include a bibliography as well as references, to demonstrate the scope of your literature search. For some secondary-source investigations, such as an annotated bibliography, the bibliography itself may be a major section of the report.

Working Scientifically Skills Outcome: BIO11/12-3

Weblink
Referencing guide
This guide is designed to help you with referencing your sources.

Weblink
Referencing i-tutorial
This tutorial will help you understand referencing and show you how to avoid plagiarism.

Information and communication technology capability

Worksheet
Writing a bibliography

Appendix

Appendices often contain information that is not essential in explaining the findings of an investigation. For example, any lengthy and repetitive information that supports your finding, such as relevant raw data, is presented in an appendix.

Worksheet
A good practical report

KEY CONCEPTS

- A formal report has the same form as an article written by a scientist. It begins with an abstract (in a scientific journal) or an aim and, at school level, background information. It includes information from a literature review on both the scientific principles behind the research and the research method selected. It includes a risk assessment, materials, method, results and analysis, discussion and conclusion. All sources need to be referenced correctly.

Other ways of communicating your work

You may want to present the results of your investigation in some other way. Scientists communicate their work in many ways. Sometimes a poster is presented or a seminar is given. An article or blog may be written or a web page created. Scientists usually use more than one means, and sometimes several means, to communicate a really interesting investigation.

Look at examples of science investigations reported on websites, in the newspaper, on the TV and so on. This will give you an idea of the different styles used in the different modes. Think about the purpose. Is it to inform, to persuade, or both? What sort of language is used?

Think about your audience and purpose, and use appropriate language and style. A poster is not usually as formal as a report. A video or web page may be more or less formal, depending on your audience.

Posters and websites use a lot of images. Images are usually more appealing than words and numbers, but they need to be relevant. Make sure they communicate the information you want them to. An example of a poster is provided on pages 30–31.

Website accessibility
The Royal Society for the Blind has information on making websites accessible.

If you are creating a website, consider accessibility. On websites, fonts need to be large enough and clear, and digital images should have tags. You can follow the weblink for more information on accessibility and web-page design.

If you make a video, then consider who your audience is and what will appeal to them. Think about how you will balance content with entertainment.

A formal report uses referencing to show where you found information. Other means of communicating about your depth study or investigation also need to acknowledge the sources of information you used. You also need to be very careful about using copyright content – for example, you cannot copy images from other people's websites unless the site's owner gives permission. Talk to your teacher about how they would like you to acknowledge your sources.

However you communicate your work, make sure you know what the message is and who the audience is. Once you have established that, you will be able to let other people know about the interesting things you have discovered in your investigation.

Ideas for depth studies

As you progress through this book, you will see investigations in each chapter. Some of these investigations are described in detail. They are designed to be useful as training exercises in how to perform primary investigations – how to set up equipment, make measurements and analyse data. Even if your depth study is from secondary sources, it is important to gain experience in doing experiments because biology is based on experiments.

At the end of each module, there is a short section called 'Depth study suggestions'. Here you will find ideas for primary- and secondary-source investigations, which build on the content of the preceding

chapters. These suggestions are sourced from experienced teachers and university academics, and from biology education literature. Your teacher will also have ideas and suggestions. You can also generate your own ideas by reading about topics you are interested in. Consider what skills from other areas you might bring to a depth study, particularly if you are artistically creative or musical. Many biologists combine their love of science with creative pursuits.

By carrying out depth studies, you will extend your knowledge and understanding in biology and, more importantly, you will learn how to work scientifically – you will learn how to do biology.

KEY CONCEPTS

- There are many ways of communicating your findings. Choose a method that is appropriate to your investigation and your intended audience.

Poster: A new fertiliser stimulates seedling growth of the newly discovered and rare *Eucalyptus pyrenea*

Arthur B. Ruse, Jacqueline Hammond, Rufus C. Smith, Karen Trace and Leigh B. Waters, Pyrenees State High School, Warrenmang, Victoria, Australia

Introduction

The recent identification of a new rare Eucalyptus species in the Pyrenees region of north-west Victoria, *Eucalyptus pyrenea* (Figure 1), has prompted efforts to try to cultivate the species (1). This would allow for regeneration of local areas with the native tree, promoting conservation of this species and enriching the natural biodiversity of the area.

Growing *E. pyrenea* seedlings has proven difficult without the addition of a fertiliser, although the standard low-phosphorus fertiliser used for *Eucalyptus* species, Super Ready, has not stimulated the same enhanced growth that is usual in other species (2).

A new fertiliser, ExtraGro, has recently become available for native Australian plants. It contains identical levels of phosphorus, potassium and trace elements to those in Super Ready but has double the amount of nitrogen, which promotes foliage growth (3). The aim of this study was to determine whether ExtraGro, containing greater nitrogen, enhanced *E. pyrenea* growth above that stimulated by Super Ready.

FIGURE 1 *An example of a Eucalyptus gum.*

Method

Genetically identical seedlings were obtained from a nursery at a length of 10 cm±2 cm and grown in 300 g of standard Australian native plant potting mixture (Figure 2). Plants were exposed to full sunlight and were watered every second day with 25 mL tap water.

Fertiliser (2 mg per seedling) was applied to the soil immediately prior to each watering.

After two weeks, seedlings were analysed for total length, shoot length, root length, total dry weight, shoot dry weight and root dry weight.

Six replicate seedlings were tested per condition.

FIGURE 2 Seedlings in their growing containers. Left pot, seedling treated with standard fertiliser (Super Ready) and right pot, ExtraGro

Results: ExtraGro stimulates enhanced root growth

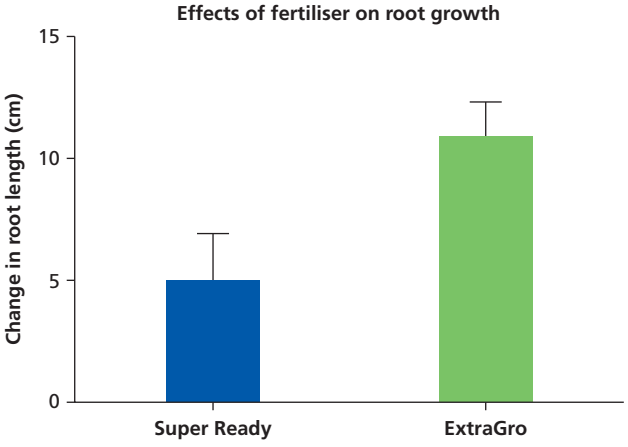

FIGURE 3 Change in root length of seedlings after two weeks of treatment, normalised to untreated seedlings (difference in treated seedling length divided by difference in untreated seedling length).

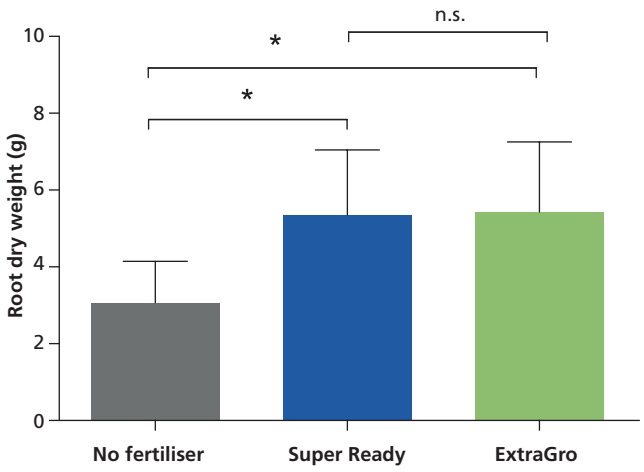

FIGURE 4 Root dry weight after two weeks of treatment. *P<0.05, n.s. = not significant. Bars show mean ± standard deviation, n = 6.

Results: ExtraGro promotes greater shoot growth

FIGURE 5 Representative seedlings after two weeks of treatment. Left, untreated seedlings. Centre, seedlings treated with Super Ready and right, seedlings treated with ExtraGro.

Sarah A. Jones, Mullan, G. D. and White, P. J. 2001. Seedling Quality: Making informed choices. Bushcare and the Department of Conservation and Land Management. Photo by G. Mullan.

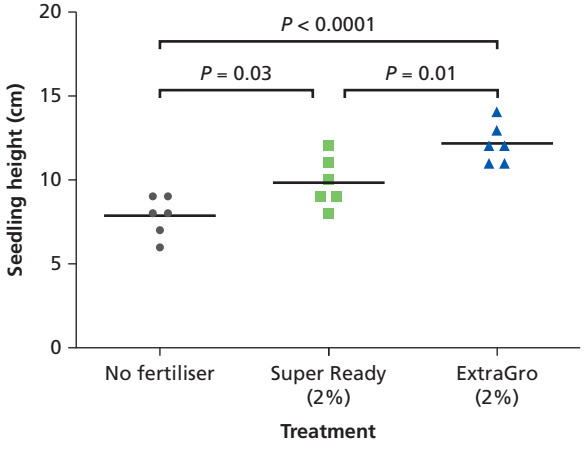

FIGURE 6 Effects of fertilisers on seedling height after two weeks of treatment

Treatment of *E. pyrenea* seedlings with the standard Australia native plant fertiliser, Super Ready, increased seedling height over a growing period of two weeks above the height reached by treatment with water alone. However, ExtraGro caused superior seedling shoot growth in terms of biomass (e.g. Figure 5) and height (Figure 6).

In addition to stimulating shoot elongation and biomass accumulation, we found ExtraGro to have superior root-enhancing properties over Super Ready. Root length increased significantly more over two weeks following treatment with ExtraGro than with Super Ready (Figure 3). However, root biomass was not increased by ExtraGro (Figure 4), suggesting that the increased nitrogen in this fertiliser stimulated the growth of longer but thinner roots.

Conclusions

We tested whether the ExtraGro fertiliser, containing twice as much nitrogen, stimulated enhanced *E. pyrenea* seedling growth over that stimulated by the standard Australian native plant fertiliser Super Ready.

ExtraGro promoted greater shoot elongation and biomass accumulation. In addition, it stimulated increased root elongation, although it did not alter root biomass gain over a two-week period.

Therefore, we have found that *E. pyrenea* seedlings respond better to a high-nitrogen fertiliser than standard fertiliser. This significant finding may inform future efforts to cultivate this species.

References

1–3. Perugia E., et al. 'A new Eucalypt native to the Victorian Pyrenees State Park'. *Australian Journal of Native Flora*, Vol 3, pp.206–8, 2011.

Acknowledgements

We thank the Pyrenees Nursery for kind provision of the *E. pyrenea* seedlings. We also thank P. Whitely for contributions to plant maintenance, and we thank our teacher Ms M Marshall for valuable contributions to our study design and poster preparation.

» MODULE FIVE

HEREDITY

2 Sexual and asexual reproduction

3 Cell replication

4 DNA and polypeptide synthesis

5 Genetic variation

6 Inheritance patterns in a population

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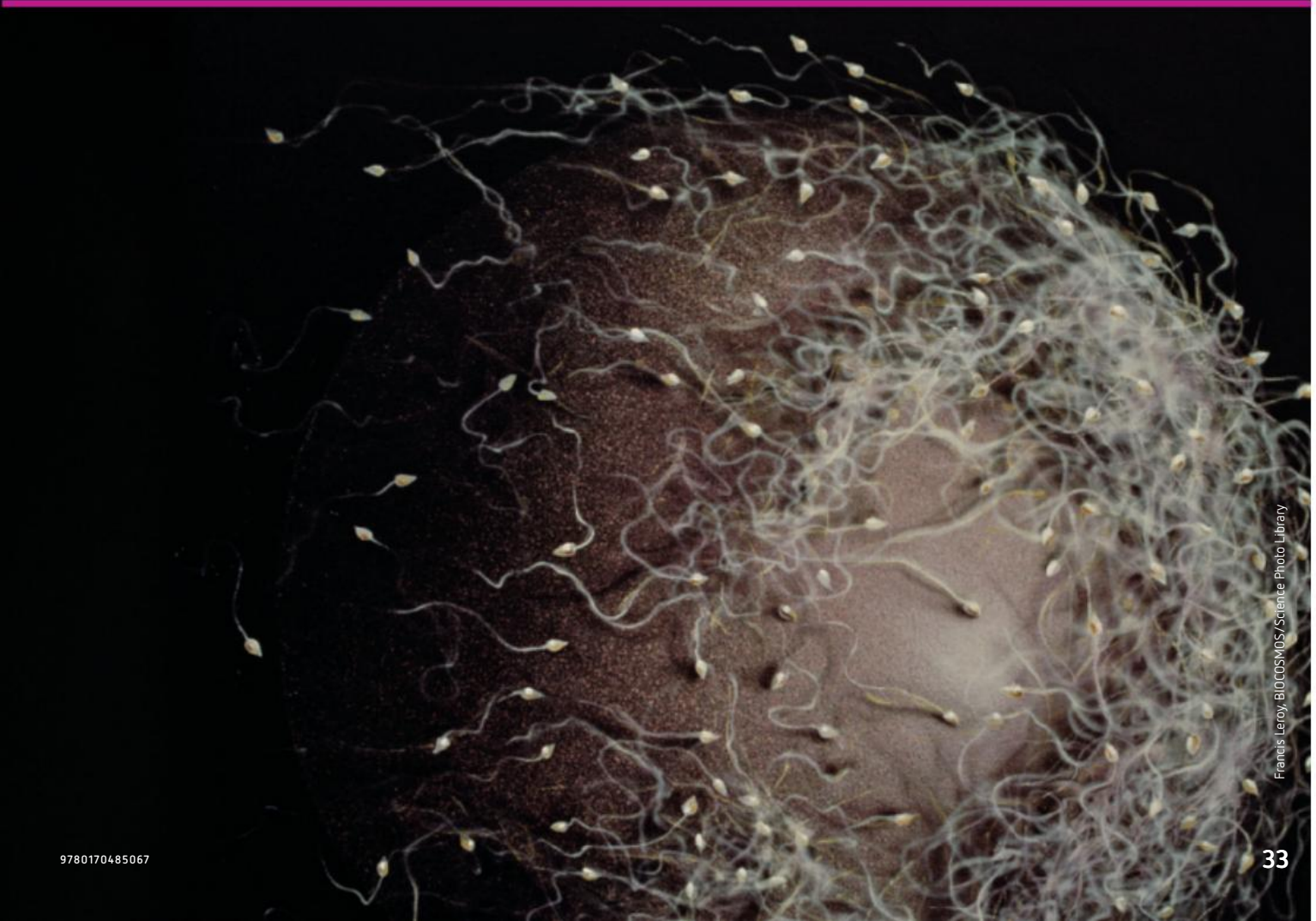
2 Sexual and asexual reproduction

INQUIRY QUESTION

How does reproduction ensure the continuity of a species?

Students:

- explain the mechanisms of reproduction that ensure the continuity of a species, by analysing sexual and asexual methods of reproduction in a variety of organisms, including but not limited to:
  - animals: advantages of external and internal fertilisation
  - plants: asexual and sexual reproduction
  - fungi: budding, spores
  - bacteria: binary fission (ACSBL075)
  - protists: binary fission, budding
- analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals (ACSBL075) CCT EU
- evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture (ACSBL074) EU L



Francis Leroy, BIOCOSMOS / Science Photo Library

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
2.1 A practical and secondary-source investigation to examine organs of perennation in plants
2.2 Practical investigation of asexual reproduction (budding, binary fission, spore formation, propagation) in living organisms
2.3 A secondary source investigation of scientific knowledge and its application to reproductive technologies in agriculture

Worksheets
- The male and female reproductive systems
- What are the social and ethical implications of applying IVF technology?
- Hormonal control of reproduction
- Human embryonic stem cell debate

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

Living organisms grow, develop, respire, feed, reproduce, excrete and eventually age and die. For continuity of life, the most critical characteristic of living organisms is reproduction. Organisms must be able to reproduce to pass on their genes, whether by replicating themselves or by mating with another individual to produce fertile offspring.

Offspring carry the same genetic traits or a mix of traits from their parents into the next generation (Fig. 2.1), ensuring that, even though individuals die, the **gene pool** and the species continue.

FIGURE 2.1 Offspring resemble their parents, physical evidence of genetic material being passed on from one generation to the next to ensure the continuation of the species.

2.1 Asexual and sexual reproduction – one parent or two?

Reproduction is a fundamental evolutionary process ensuring the continuity of life. Considering the origin of life and the evolution of living organisms, reproduction must have been one of the first characteristics of life to arise. The ability of a chemical system to make copies of itself and, later, the ability of an organism to make copies of itself, are of primary importance in ensuring the continuity of species.

The **reproductive success** of an organism is determined by its ability to produce fertile offspring that survive to reproductive maturity and produce offspring of their own, in this way replacing the parent. **Biological fitness** is a measure of an individual's reproductive success. Biological fitness is calculated as the average contribution to the gene pool made by a certain genotype within a population and the relative likelihood that those **alleles** (variants of a gene) will be represented in future generations. An allele with higher fitness is more likely to be represented in future generations than an allele of the same gene with lower fitness. Reproductive success is a feature of an individual, while biological fitness is a feature of an allele in a population.

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There are two main methods of reproduction. **Asexual reproduction** involves only one parent and gives rise to offspring that are genetically identical to each other and to the original parent.

Sexual reproduction usually involves two parents who produce offspring that have a mix of the parents' genes and therefore differ from each other and from the parents. (Occasionally, organisms are bisexual and if self-fertilisation occurs, the offspring would arise by sexual reproduction from one parent.)

KEY CONCEPTS

- Reproduction means making a copy or a likeness. For living organisms, this means producing offspring that are identical to the parent or resemble the two parents that gave rise to them.
- Individuals have a finite lifespan, so in order for a population or a species to survive, genetic material must be passed from one generation to the next. This ability to reproduce is known as the reproductive success of an individual.
- The genetic material of all organisms in a population makes up the gene pool. The likelihood of genes appearing in the next generation and being passed on is known as biological fitness.
- In evolutionary terms, reproduction is less significant for individual success and more important for the continuation of the species.

Advantages of sexual reproduction

Sexual reproduction involves the meeting of special sex cells called gametes, which carry genetic information from both parents to the offspring. As a result, the offspring contain a mix of parental genes and are not genetically identical to the parents or to other offspring, and this introduces genetic variation into the population. The greatest advantage that sexual reproduction is thought to provide, in terms of continuity of life at the species level, is genetic diversity.

Some offspring may possess random variations that make them better suited to new and changing environmental conditions. They may out-compete their parents and/or other individuals in the population, thereby gaining a selective advantage. In the face of environmental change, the survival of some individuals gives the overall population or species a better chance of survival.

The disadvantage of sexual reproduction is that this process demands a greater expenditure of time and energy, involving processes such as finding a mate, courtship behaviour, gamete production and mating, before the production of young. These processes may also make organisms more vulnerable to predators. Because sexual reproduction requires far more investment by an individual than asexual reproduction, it tends to be the first functional process that is sacrificed in times of hardship.

Sexual reproduction – the meeting of two gametes

During sexual reproduction, a combination of genetic material from two parents is passed on to offspring. Every species has a characteristic number of chromosomes per cell. For example, humans have 46 chromosomes, camels have 70, tomatoes have 24 and chickens have 78. Each species usually has two sets of chromosomes, arranged in homologous pairs. The number of chromosomes does not necessarily reflect the complexity of the organism and even varies among closely related species. For example, the housefly has 12 chromosomes, whereas the fruit fly has only 8. The important thing to remember for studies of genetics is that the chromosome number is constant for each species and does not change from one generation to the next.

In sexual reproduction, to prevent the chromosome number from doubling in each successive generation, a mechanism to ensure that each parent contributes only half of his or her chromosomes to their offspring is necessary. **Meiosis**, a type of cell division that takes place in the reproductive organs of plants and animals, is important to maintain the characteristic chromosome number during sexual reproduction. When a cell involved in sexual reproduction divides by meiosis to produce **gametes** (sex cells), the chromosome number halves – that is, each resulting gamete contains only *one set* of chromosomes.

The terms **diploid** and **haploid** refer to the number of *sets* of chromosomes within any cell. In most organisms, the **somatic cells** (body or non-reproductive cells) contain *two sets* of chromosomes – that is, the diploid number of chromosomes (in humans this number is 46 or 23 pairs).

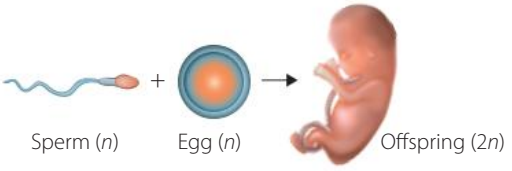

FIGURE 2.2 The sperm and the egg are haploid gametes that give rise to diploid offspring in sexual reproduction.

Offspring inherit one set of chromosomes from the mother (**maternal** chromosomes) and one set from the father (**paternal** chromosomes) (Fig. 2.2). A fertilised egg or **zygote** arises as a result of the fusion of haploid gametes, when the chromosome number changes from haploid to diploid. The diploid zygote divides by mitosis to become an embryo with identical body cells that all have the diploid number of chromosomes.

If $n = 1$ set of chromosomes, then in humans, $n = 23$. Human somatic cells are diploid ($2n$) and have 46 chromosomes, whereas human gametes are haploid (n) and have 23 chromosomes (as a result of meiosis). Fertilisation of an egg by a sperm restores the diploid number (Fig. 2.3).

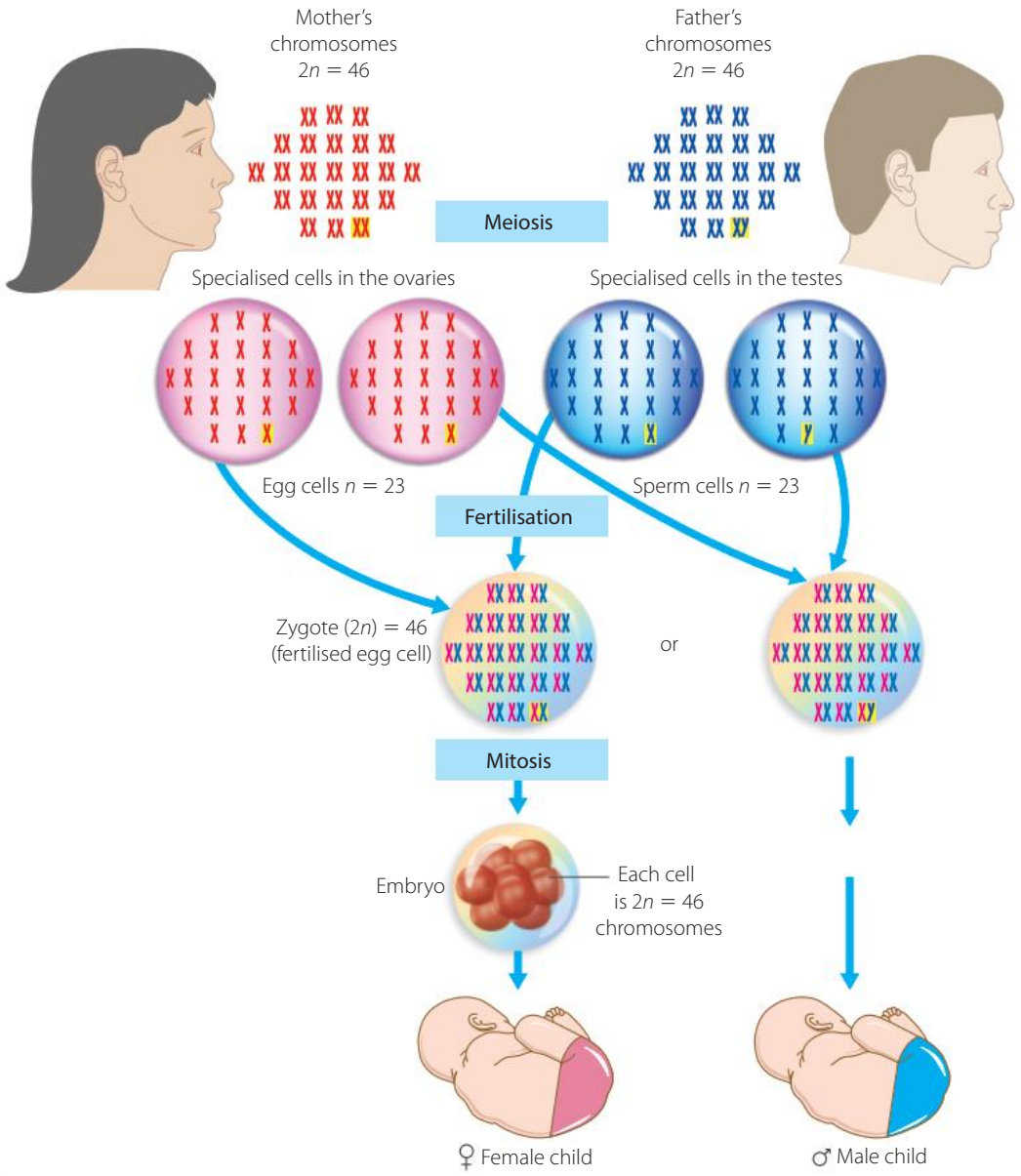

FIGURE 2.3 Maintenance of the diploid number during sexual reproduction in humans

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KEY CONCEPTS

- Sexual reproduction requires the production of male and female gametes (sperm and ova) by the process of meiosis (reduction division).
- Each gamete is haploid (n) – that is, it has half the normal number of chromosomes.
- The gametes fuse during the process of fertilisation to create a zygote (fertilised egg) with the full diploid ($2n$) complement of chromosomes.
- In offspring, 50% of the chromosomes come from the mother and 50% from the father.
- The cells of the zygote divide by mitosis, keeping the chromosome number constant, and the resulting embryo continues to grow and mature into a new individual.
- Fertilisation and meiosis are reciprocal processes – that is, one is a fusion from haploid to diploid, and the other is a reduction from diploid to haploid.

CHECK YOUR UNDERSTANDING 2.1a

1. Explain what is meant by each of the following terms: diploid, haploid, gamete, somatic cell, paternal, maternal.
2. Distinguish between asexual and sexual reproduction.
3. Define 'reproductive success' and 'biological fitness'. What is the difference between these two terms?
4. Give one advantage and one disadvantage of sexual reproduction.
5. What is meiosis? Why is this process important to a species?
6. What is the importance of variation in a population?

Sexual reproduction in animals

Sexual reproduction is a mechanism that has evolved to ensure continuity of species. In animals, a number of sexual reproductive strategies ensure that reproduction occurs effectively in the environment in which an organism lives.

Most animals are unisexual – there are separate male and female individuals. However, a small range of animals are bisexual or **hermaphrodites**, where each individual has both male and female reproductive organs. Hermaphroditism can be advantageous to species with low population densities, or in animals that are non-motile (such as coral), where finding a mate is difficult. The disadvantages of hermaphroditism are that individuals must expend larger amounts of energy to grow and maintain two sets of reproductive organs. Then if self-fertilisation occurs, the gametes carry fewer possible combinations of genes and therefore the offspring will have less variation.

Other reproductive strategies include the type of fertilisation (internal or external), the number of gametes produced, the timing of gamete release, where the young develop (outside or inside the body) and the nature of parental care. If these strategies are advantageous within the particular environment in which an organism lives, they can increase its reproductive success.

Fertilisation – external or internal?

In animals, the union of male and female gametes (sperm and ova) can occur outside the body (**external fertilisation**) or inside the body (**internal fertilisation**). The key to successful fertilisation of ova by sperm is that the gametes, each of which is a single haploid cell surrounded by a cell membrane, must meet and not dehydrate in the process. Therefore, external fertilisation is better suited to organisms that reproduce in an aquatic environment (such as marine creatures) or a very moist environment (such as earthworms), whereas internal fertilisation is typical of many terrestrial organisms (such as insects, lizards and kangaroos). Vertebrate sexual reproduction is thought to have started in the ocean (fish) and freshwater environments (amphibians) and then evolved once vertebrates such as reptiles, birds and mammals colonised the land and the air. The change in type of fertilisation (external or internal) is consistent with the accepted sequence of species' evolution.

Invertebrates are animals without a backbone, such as coral polyps, insects and snails.

Vertebrates are animals with a backbone, such as fish, amphibians, reptiles, birds and mammals.

The chances of successful external fertilisation are increased by synchronisation of reproductive cycles, mating behaviours and the release of gametes. When fertilisation and development of the young take place externally, there is little or no parental care. This means that less time and energy are required of the parents, but a larger number of gametes must be produced to ensure that some young survive. The advantage of external fertilisation is the wide dispersal of young. Some marine animals release their gametes into the sea, and fertilised eggs are carried away to settle in an area far from their parents. This reduces competition for food and living space, and also allows rapid recovery of populations away from damaged areas.

Staghorn coral

Pheromones are chemical substances released by one organism that have an effect on another organism.

Staghorn coral is an example of a colony of invertebrate marine animals (polyps) that achieve fertilisation by simply shedding millions of gametes into the sea (Fig. 2.4). Environmental cues, such as water temperature, tides and day length, help synchronise the reproductive cycle. When polyps in one coral colony start to spawn, **pheromones** released along with gametes stimulate nearby individuals to spawn, resulting in coordinated spawning over a wide area.

FIGURE 2.4 Staghorn coral (*Acropora yongei* sp.) releases bundles containing sperm and eggs.

During the mass spawnings of coral on Australia's Great Barrier Reef, the number of gametes shed is so great that, for a time, the sea turns milky. Within one day, fertilised eggs develop into swimming larvae. After a few days at the surface, the larvae descend to find a suitable site to form a new colony. Although millions of staghorn coral larvae are produced, almost all are eaten by predators. Of the few remaining, only a tiny proportion reach adulthood.

Bony fish

The females of most species of marine bony fish produce eggs (ova) in large batches and release them into the water, where they fuse with sperm outside the body of the female. Because the gametes disperse quickly, the release of large numbers of eggs and sperm from the females and males must occur almost simultaneously. In most marine fish, the release of gametes is restricted to a few brief and clearly determined periods. Although thousands of eggs are fertilised in a single mating of bony fish, many of the resulting offspring succumb to microbial infections or predation, and few survive to maturity.

FIGURE 2.5 Copulation in frogs, where eggs are fertilised externally

Amphibians

Amphibians invaded the land without fully adapting to the terrestrial environment, and so their life cycles still involve stages in water. Gametes from both males and females are released in fresh water, such as ponds or streams. In frog and toad copulation, the male grasps the female and straddles her back, discharging fluid containing sperm onto the eggs as they are released by the female into the water (Fig. 2.5).

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An enormous number of gametes are produced by amphibians, first to ensure that many undergo fertilisation, and second to ensure the production of a large number of offspring. Because most amphibians provide no parental care, the young tadpoles are easy prey and not many survive to reproductive age.

FIGURE 2.6 A young froglet emerging from the mouth of a female Southern gastric brooding frog (*Rheobatrachus silus*) after developing in its mother's stomach

Some frogs, such as the Southern gastric brooding frog (discovered in forests north of Brisbane in 1974 and recently registered as extinct), evolved special adaptations to ensure survival of the young. The eggs of the terrestrial frog were fertilised by sperm externally in a watery environment, after which the female would swallow the eggs, and the young developed internally in the female's stomach. In the stomach, digestive secretions ceased and the eggs settled into the stomach wall, where they were protected and absorbed nutrients from the mother for about 6–7 weeks (during which time the female did not eat). Young frogs were then regurgitated through the mouth (Fig. 2.6). This mechanism provided some protection for the underdeveloped young from predation, infection and dispersal, significantly increasing the chance of successful survival of the offspring. These animals were unusual in having external fertilisation but internal development, and provide an extreme example of parental care.

Internal fertilisation and parental care in animals

Organisms that undergo internal fertilisation tend to be adapted to terrestrial environments and reproduce successfully on land. The internal environment for fertilisation not only protects gametes from dehydration and loss to external elements, but also protects the fertilised eggs and developing young from immediate predation. Therefore, with internal fertilisation, fewer eggs are required for the survival of a sufficient number of offspring. The internally fertilised egg may develop a shell and be laid in the external environment (**oviparous**) to complete its development (in reptiles and birds, for example), or it may continue to develop inside the female's body.

In most mammals, the fertilised egg becomes an embryo that is nurtured inside the female parent's body, obtaining nutrients through a placenta, and is born alive (**viviparous** development). In rare instances, a combination of the above occurs and eggs with yolk for nourishment are retained inside the mother's body until they are ready to hatch. Newly hatched young are born alive (**ovo-viviparous**, for example in some snakes and sharks).

FIGURE 2.7 Reptiles such as tortoises copulate, so the eggs are fertilised internally.

Reptiles

Most reptile eggs are fertilised internally and then deposited outside the mother's body for development. During copulation (Fig. 2.7), male reptiles use a tubular penis to introduce sperm into the female.

FIGURE 2.8 After developing in a yolky egg, tiny crocodiles (*Crocodylus porosus*) hatch.

In crocodiles, fertilisation occurs internally. The female crocodile (*Crocodylus porosus*) lays small numbers of large yolky eggs in clutches along the sandbanks beside the sea or a river (Fig. 2.8). The eggs of most reptiles are covered in a soft but tough leathery shell. Exceptions are tortoises, geckos and crocodiles, which lay hard-shelled eggs. The eggs contain sufficient food reserves to last until the eggs hatch. The offspring resemble miniature adults and are able to crawl from the buried nest to the surface and make their way to water, a journey that makes them vulnerable to predation. There is no parental care of these young.

It interesting to note that the temperature at which reptile eggs are incubated often determines whether the resulting individuals are male or female. At high temperatures, females hatch, even from eggs in which the individual has male chromosomes. These females are able to lay eggs and reproduce like normal females, despite being genetically male.

Birds

Courtship behaviour by birds may take place in flight or on the ground, but copulation takes place on the ground, making the birds vulnerable to predators. Fertilisation is internal. Most male birds do not have a penis, so during copulation, male and female birds rub the openings of their **cloacas** together and sperm are transferred to the female's body. In some larger birds (such as swans) the male cloaca extends to form a 'false' penis. Once fertilised, the ovum passes along the oviduct and successive glands secrete yolk, followed by protein (albumen, commonly called egg white) around it. A calcium carbonate shell is then secreted and this hardens when the egg comes into contact with the air, directly after it is laid. The hard shell distinguishes bird eggs from soft-shelled reptilian eggs and gives more protection. Most birds incubate their eggs after laying them, to keep them warm, and exhibit parental care once the young have hatched.

Monotremes are mammals, such as the platypus and the echidna, that lay eggs but still suckle their young.

FIGURE 2.9 A young echidna with its mother

Mammals

The gametes of all mammals undergo fertilisation internally. Mammals are divided into three subclasses – monotremes, marsupials and eutherians – based on the subsequent development of their embryos.

Monotremes, such as the platypus and the echidna, are oviparous. After internal fertilisation, they lay eggs that develop outside the mother's body. Platypus parents incubate their eggs in a nest, whereas echidnas place their eggs into an abdominal pouch where they stay for about seven weeks. The young hatchlings (puggles) obtain milk from their mother's mammary glands by licking her abdominal skin (Fig. 2.9).

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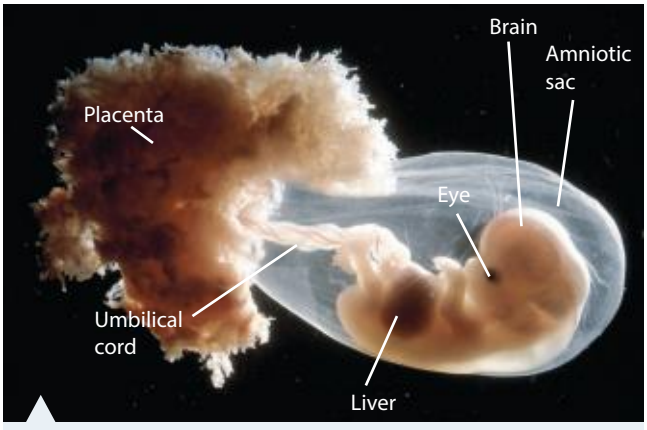
Young **marsupials** develop internally for a short time after fertilisation and then continue their embryonic development in a pouch. Offspring are born at a very young age and crawl up the mother's abdomen to the pouch (Fig. 2.10). In favourable environmental conditions, marsupials such as the kangaroo can have three offspring at different stages of development at any one time – one out of the pouch but still drinking milk, one in the pouch attached to a nipple, and a fertilised ovum at the blastocyst (ball of cells) stage in the uterus. The development of the youngest is triggered when the second-youngest detaches from the nipple and leaves the pouch. Because milk production in kangaroos lasts much longer than a pregnancy, it is necessary to delay development of the new embryo until the older one is no longer suckling. This delay in development, termed **embryonic diapause**, is another strategy to increase chances of survival.


Jean-Paul Ferrero/ AUSCAPE/ Alamy Stock Photo

FIGURE 2.10 Kangaroos give birth to small foetuses, which complete their development in a pouch.

The red kangaroo has a special reproductive adaptation to ensure survival of the young in times of drought. If the mother is unable to produce sufficient milk to sustain a joey attached to a nipple in the pouch, this joey dies and a newborn individual will enter the pouch a month later. The tiny newborn requires far less milk for the first few weeks in the pouch. This strategy ensures the continuity of young who are ready for development when the drought ends and allows very rapid population growth when conditions are good. However, in prolonged drought conditions, kangaroos stop breeding and only begin again when rain triggers a hormonal response in the female. This very effective mechanism restricts reproduction to times when conditions favour survival of the young.

Eutherians, otherwise known as placental mammals, include dingoes, rodents (such as rabbits, rats and mice), domesticated animals (such as dogs, sheep and cattle) and humans. Following internal fertilisation, the young completes its embryonic development inside the body of the mother in a special organ, the uterus, which nurtures and protects the embryo. Once one or more fertilised eggs implant into the uterine wall, a placenta develops, connecting the young to a supply of nutrients and oxygen that passes from the bloodstream of the mother to the developing young (Fig. 2.11). Excretory wastes such as nitrogenous wastes and carbon dioxide from the embryo diffuse across the placenta to the mother's body, where they are excreted along with the mother's own wastes. The mother gives birth to live young that are mature and therefore have a greater chance of survival. This type of development, where live young are born, is described as viviparous. Placental mammals produce one to a few young at a given time and they invest a large amount of energy in parental care, increasing the chance of survival of the young.



Leemage/Corbis Historical/Getty Images

FIGURE 2.11 Human embryo developing in a uterus attached to a placenta

Internal and external fertilisation in animals are compared in Table 2.1.

TABLE 2.1 Comparison of internal and external fertilisation in animals

CHARACTERISTICS	DIFFERENCES		SIMILARITIES
	EXTERNAL FERTILISATION	INTERNAL FERTILISATION	
Gametes	Large numbers of male and female gametes produced	Large number of male gametes and fewer female gametes produced	Male and female gametes required – sperm and eggs (ova)
Union	Occurs in open water environments	Occurs inside the reproductive tract of the female in organisms that live mostly or completely on land	Sperm fertilise the eggs when they unite
Conception mechanism	Simultaneous release of gametes	Copulation: the male inserts sperm into the female's reproductive tract via penis or cloaca	Sperm will fertilise eggs when in very close proximity to each other; gametes require a watery environment for this to occur
Chance of fertilisation	Low, because male gametes are released into a large open area where there is less chance of successfully uniting with female gametes	High, because male gametes are released into a confined space where there is more chance of successfully uniting with female gametes	If male and female gametes are in close proximity to each other, fertilisation will usually occur
Environment for zygote	Usually external, in a watery environment that is vulnerable to environmental elements such as temperature, predation, infection and rapid dispersal from the area	Usually internal, in a very protected environment inside the female's body. Temperature is controlled and there is less chance of predation, infection and loss of zygote from the area	Zygote requires a watery environment for development
Number of offspring/zygotes	Usually a larger number than in internal fertilisation, but many zygotes perish and so a smaller number of offspring survive	A smaller number of offspring than in external fertilisation, because very few perish (higher success rate)	Zygote number is determined by the number of sperm and ova that successfully fuse
Breeding frequency	More frequent than in internal fertilisation due to the lower fertilisation success rate	Seasonal and less frequent than in external fertilisation due to higher fertilisation success rate and greater energy costs	Breeding frequency depends on the requirements of the species and the favourability of environmental conditions
Parental investment	Usually no parental care	Parental care of eggs and/or developing young is more common	Parental investment is indirectly proportional to the number of gametes produced

KEY CONCEPTS

- Fertilisation is the union of male and female gametes and may take place externally or internally.
- External fertilisation occurs in aquatic or moist terrestrial environments, to prevent dehydration of gametes. Gametes must be produced in large numbers to ensure success.
- Internal fertilisation takes place inside the body of the female and involves mate attraction and copulation, which require energy investment and put the organisms at risk of predation, but fewer eggs need to be produced.
- External fertilisation occurs in most invertebrates and some vertebrates (fish and amphibians).
- Internal fertilisation occurs in some invertebrates (insects and snails) and most vertebrates (reptiles, mammals and birds).
- Other mechanisms that increase the chances of survival and continuity of species include nourishment for the developing young and parental care.

CHECK YOUR UNDERSTANDING 2.1b

1. Describe the conditions under which asexual reproduction is advantageous, using a named example.
2. What are the advantages and disadvantages of sexual reproduction?
3. Give examples of one vertebrate and one invertebrate that have external fertilisation. Explain how they ensure the gametes meet.
4. Give examples of one vertebrate and one invertebrate that have internal fertilisation. Describe the habitat in which they live.
5. Describe the features of internal and external fertilisation that are similar and those that are different.
6. Give two advantages and two disadvantages of internal and external fertilisation.

Sexual reproduction in plants

Sexual reproduction in plants also relies on the successful fusion of male and female gametes. However, in plants, this fusion is more difficult because plants grow in the ground and cannot move. The broad range of plants on Earth, from less advanced plants such as mosses and ferns to cone-bearing plants (gymnosperms) and flowering plants (angiosperms), have developed a range of reproductive strategies to ensure the continuity of species. These strategies include relying on external agents to carry the gametes from one parent to another, commonly called pollinating agents. Plants also rely on external agents to disperse their seeds (wind, water and animals, for example) and, because they do not have the ability to move away from extreme heat or cold, they have other survival strategies for seeds once they land.

As early angiosperms evolved, advantageous features in flowers that resulted in more successful pollination would have been selected by natural means, and these features would have been retained within populations, leading to the great diversity of flowering plants today.

To understand the process of sexual reproduction in plants, it is necessary to examine the structure of flowers, which are the reproductive organs of plants. A flower contains either female reproductive parts (carpel or gynoecium), male reproductive parts (stamens) or both, in addition to their non-sexual parts (petals and sepals) (Fig. 2.12).

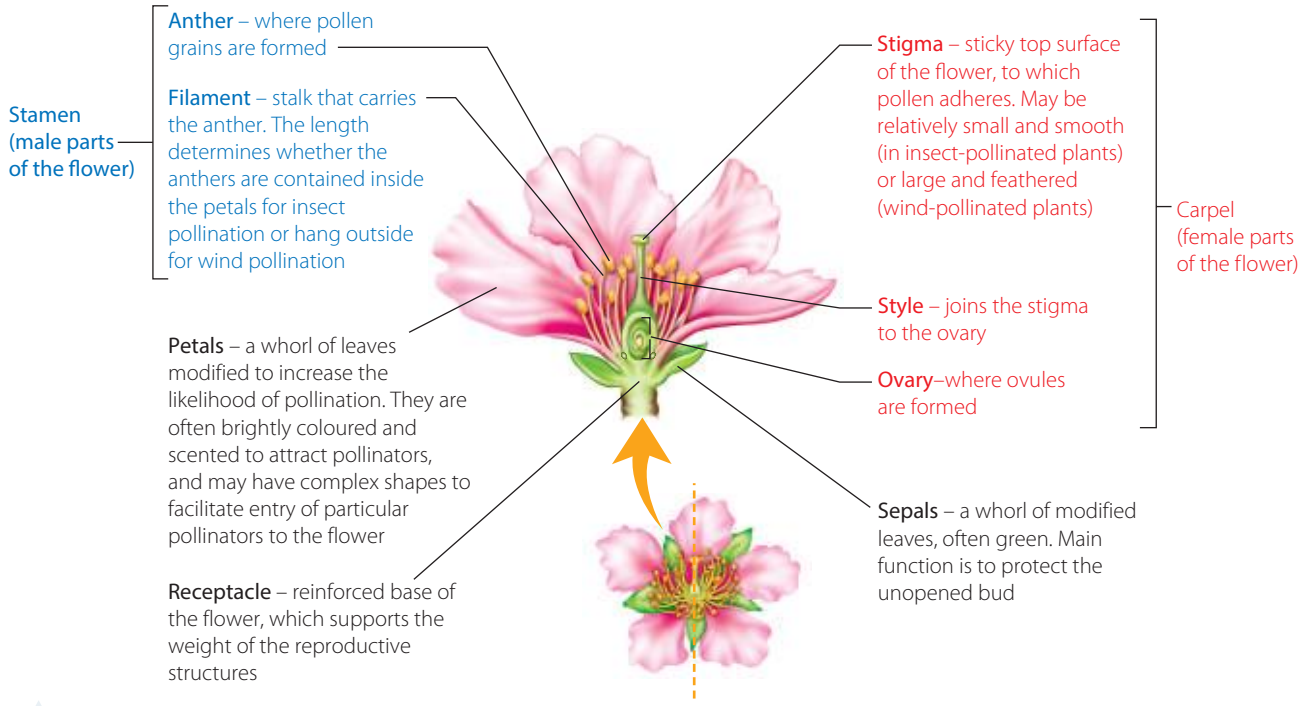


FIGURE 2.12 Top view and longitudinal section through a flower (male parts labelled in blue and female parts in red)

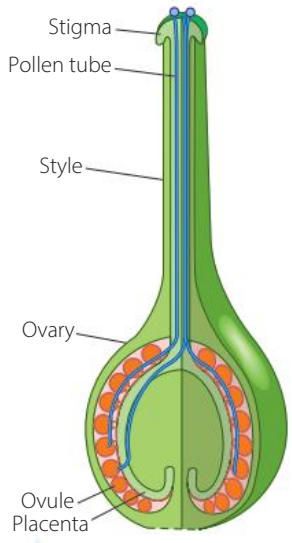


FIGURE 2.13 The pathway taken by a pollen tube as it grows from the stigma to the ovary

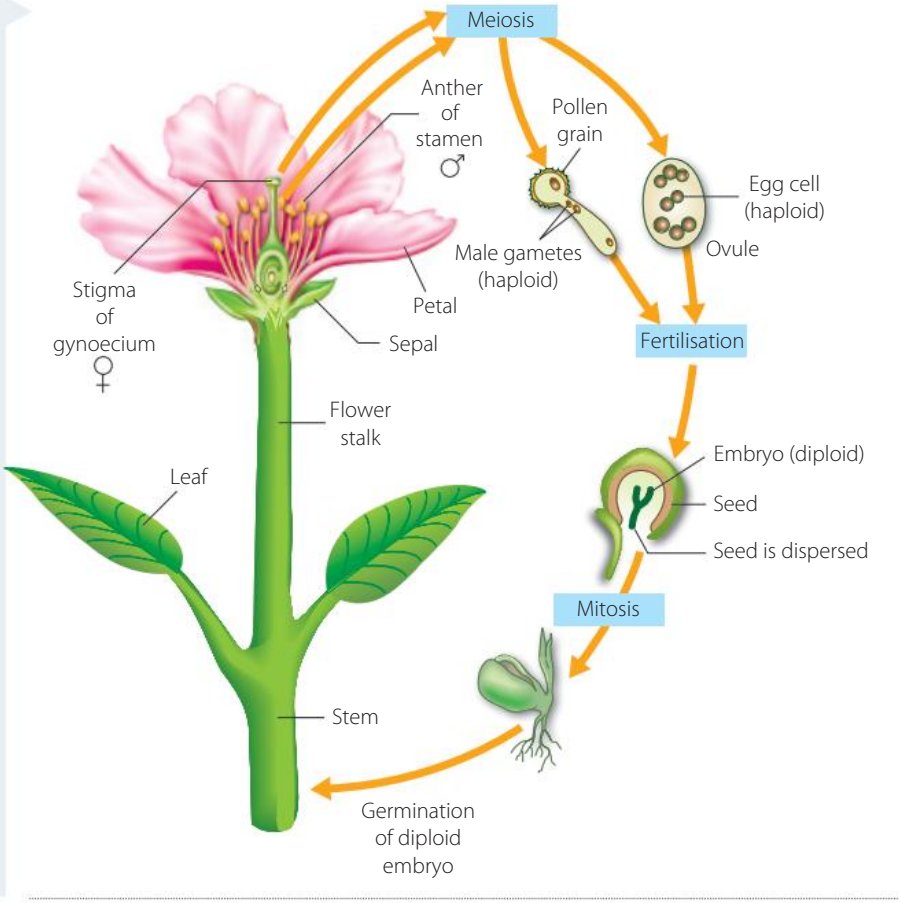
In order for fertilisation to occur, the male gametes inside **pollen** must be carried from the **anthers** to the female part of a flower, called the **stigma**. This process of gamete transfer is called **pollination**. Once pollen has been deposited on the stigma, a pollen tube germinates and grows down the style, carrying inside it the male gamete (sperm cell) to an **ovule** contained in the **ovary** (Fig. 2.13). In flowering plants, fertilisation occurs internally inside the ovary.

Pollination, fertilisation and seed production

Plants are dependent on agents such as wind, water and animals to carry their pollen, from the anthers of one flower to the stigma of a flower either on another plant (cross-pollination), or on the same plant (self-pollination). Cross-pollination ensures greater variation in the offspring. A strategy that is seen in some plants to favour cross-pollination rather than self-pollination is for the plant's pollen to mature at a different time than its stigma.

During fertilisation, the sperm cell that was transferred by the pollen tube fuses with the egg cell (ovum) inside the ovule in the female part of the flower. The fertilised ovule develops, protected within the ovary. The ovule containing an embryo is now termed a **seed** and the surrounding ovary grows to become a **fruit** (Fig 2.14).

FIGURE 2.14 Sexual reproduction life cycle in flowering plants



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Self-pollination versus cross-pollination

In plants, self-pollination requires less energy as there is no requirement for the plant to produce structures to attract pollinators, such as brightly coloured petals or nectar. These plants can grow in areas where insects or other animals that visit plants are absent or very few in number.

Plants that undergo cross-pollination rely on outside agents to transfer pollen from anthers to stigmas. These may be abiotic agents such as wind or water, or biotic agents such as insects, birds and mammals. As flowers become increasingly specialised, so do their relationships with pollinating agents.

Pollination by wind

Many angiosperms are wind pollinated and their flowers are small, greenish and odourless, with reduced or absent petals. The flowers are grouped together in fairly large numbers and may hang down in tassels that blow around in the wind and shed pollen freely (Fig. 2.15). Many Australian grasses are wind pollinated. In these species the anthers are very long and produce large amounts of light pollen, which is easily picked up by the wind passing over the flowers. Usually the stigmas are also very large and spread out in a feathery manner to trap pollen carried by wind.

Wind pollination is very inefficient, so large quantities of pollen are produced. Different pollen grain structures between species ensure compatibility within the same species.

NNehring/E+/Getty Images

FIGURE 2.15 Small, wind-pollinated grass flowers (*Lolium perenne*)

Pollination by animals

Flowers that attract animals are more effective in ensuring the transfer of pollen and, therefore, the reproductive success of that species. Special adaptations in the flower are of considerable advantage, because a one-to-one relationship between a plant and an animal species reduces wastage of pollen by ensuring that it is deposited on the correct flower. Animals such as insects, birds and mammals that act as pollinators search in flowers for a reward, such as a meal of nectar (a sugary liquid secreted by nectaries in the flower) or pollen. During this search, pollen rubs onto their bodies and is then transferred to the next flower they visit. Flower scent, colour, markings, shape and nectar are important in attracting animals, all differing between each flower species and adapted to the type of animal they attract (Table 2.2, Fig. 2.16).

TABLE 2.2 Comparison of wind-, bird- and insect-pollinated flowers

FEATURE OF FLOWER	WIND-POLLINATED FLOWERS	BIRD-POLLINATED FLOWERS	INSECT-POLLINATED FLOWERS
Petals	Small and inconspicuous, usually green or dull in colour	Usually large and colourful, red or orange, often a tubular shape, sometimes no petals at all	Usually large and colourful (yellow or blue); may be shaped to encourage specific pollinators
Scent	Usually absent	Rarely fragrant because birds have little sense of smell	Often present because insects are highly attracted to scents
Nectar	None	Large amounts of nectar produced in nectary at base of flower	Sometimes produced at base of petals, so insect must enter the flower to reach the nectar
Anthers	Anthers protrude outside the flower, so pollen is easily blown off by the wind; abundant pollen is produced	Anthers are commonly lower than stigma, colourful, and may not be enclosed by petals	Enclosed within flower, commonly lower than stigma
Stigma	Stigma protrudes from the flower, is often long, feathery and sticky, to increase surface area for trapping wind-borne pollen	Higher than anthers, sometimes not enclosed by petals and often colourful	Enclosed within flower, sticky, and commonly higher than the anthers
Pollen	Very small grains, light and powdery; large amounts produced	Sticky or powdery pollen; small amount produced	Relatively large grains and often sticky; small amount produced

Some Australian flowers (*Callistemon* – bottlebrush, *Banksia* and *Grevillea* species) are pollinated by small mammals such as bats, possums, pygmy possums and small rodents (Fig. 2.16).

FIGURE 2.16 Examples of native animals and insects pollinating flowers; **a** the red colour of the New South Wales waratah (*Telopea speciosissima*) attracts birds as pollinators; **b** a blue-banded bee pollinating a purple flower (bees cannot see red; they are attracted to flowers in the blue and yellow range); **c** a hammer orchid (*Drakaea glyptodon*) mimics a female wasp; male wasps act out mating behaviour with the orchid flower, effecting pollination; **d** the Australian honey possum (*Tarsipes rostratus*) pollinates flowers while feeding on nectar.

Seed dispersal

After pollination and fertilisation of the flowers of a plant, seeds (fertilised ovules) from inside the ovary are dispersed (Fig. 2.17). It is an advantage for seeds to be dispersed over a wide distance, as this helps prevent overcrowding and competition for light, water and soil nutrients. Widespread distribution also increases the chances of continuity of the species in other locations, in case there is a sudden change in the local environment, such as fire or disease. Australian native plants have evolved a variety of adaptations to increase the chance of successful dispersal of their seeds.

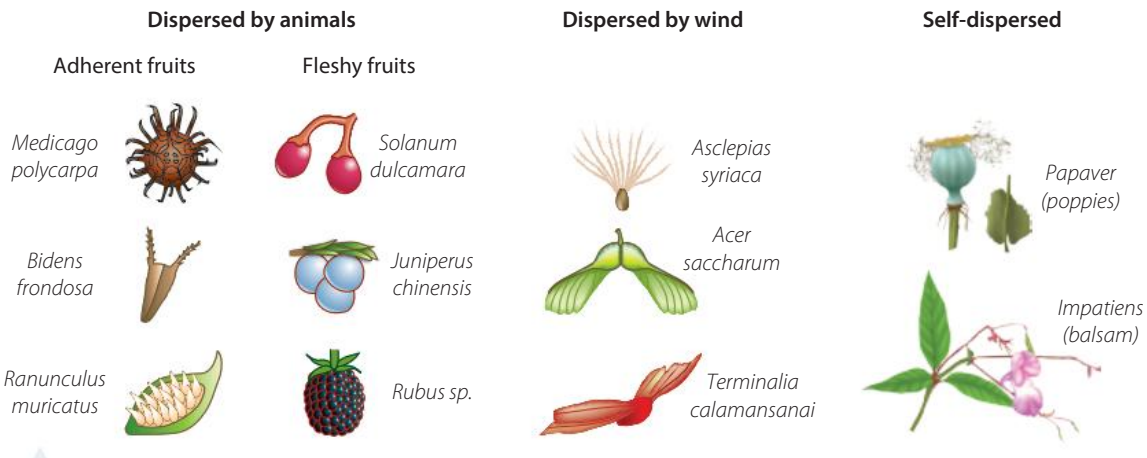

FIGURE 2.17 Adaptations of different types of seeds to facilitate their dispersal

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The success of seed dispersal depends on the type of agent that the plant relies on. Fruits may be dry (such as banksia pods, gum nuts) or fleshy (such as apples, dragon fruit) (Fig. 2.18). Dry fruits often have inbuilt 'explosive' mechanisms for dispersal by air, wind or water (abiotic agents). They are usually light so that they can float on air or water. Fleshy fruits often rely on insects, birds or mammals for dispersal (biotic agents) – the animals eat the fruit, move along and then egest the seeds, usually some distance away from the parent plant.

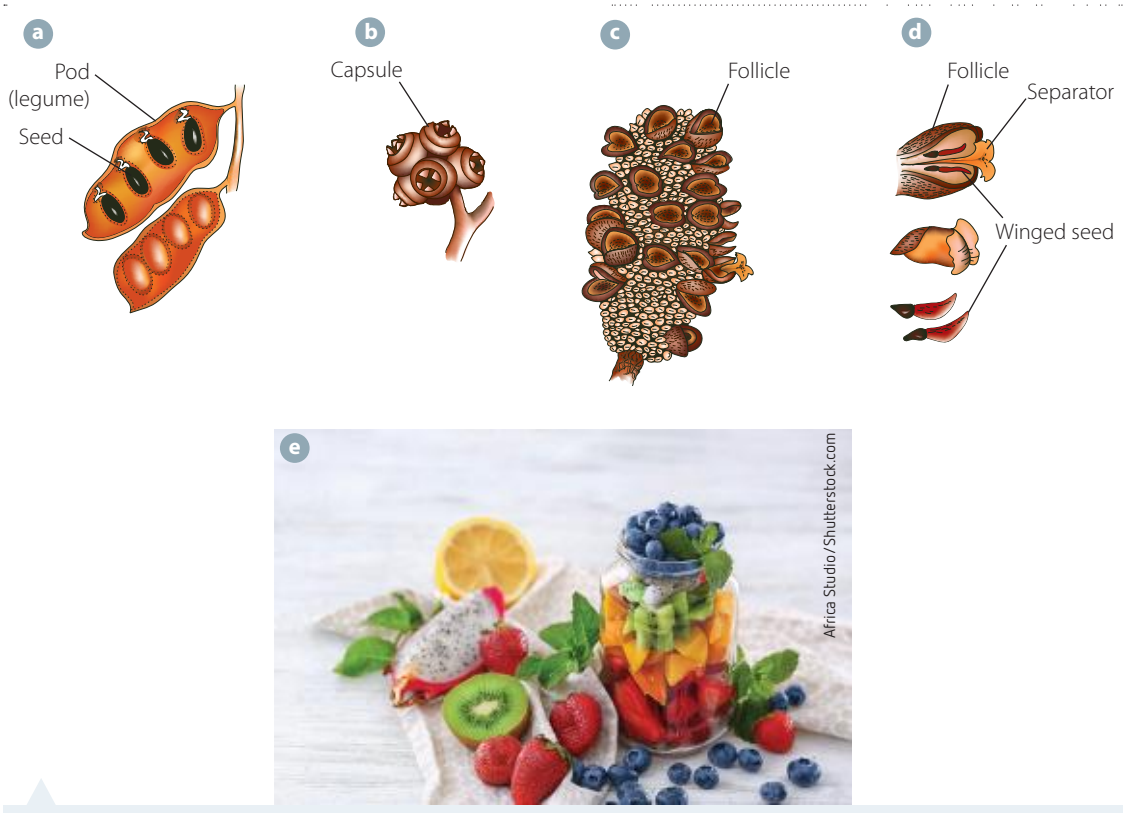

FIGURE 2.18 Examples of dry and fleshy fruits: **a** pods or legumes of *Acacia*; **b** capsules of *Eucalyptus*; **c** follicles of *Banksia*; **d** follicle separator (*Banksia*) – when wet it pulls the two seeds out of the fruit; **e** the seeds of fleshy fruits such as strawberries, blueberries, kiwifruit and dragonfruit are contained within a fleshy ovary (fruit).

Germination

The plant embryo inside a seed is in a dehydrated form and is dormant, to allow the seed to survive adverse conditions. If the seed lands in suitable soil that provides sufficient water, oxygen and warmth, it germinates – that is, the embryo begins to grow, producing a **radicle** or young root to absorb water and soil nutrients, as well as a **plumule** or young stem, which develops green leaves for food production by photosynthesis. Once the seedling becomes established, it grows and develops into an adult plant that can begin the reproductive cycle once again.

KEY CONCEPTS

- Sexual reproduction in plants involves external agents of pollination and seed dispersal, such as wind, water, fire (abiotic) and animals (biotic).
- Pollination mechanisms in plants include self-pollination (to ensure survival if reproductive partners are scarce) and cross-pollination (to increase genetic variation and ensure survival if a sudden environmental change such as disease or drought occurs).
- The life cycle of a plant involves pollination, fertilisation, seed dispersal and germination.
- Seed dispersal relies on the type of fruit in which the seeds occur matching the type of dispersal agent available in that environment.

- Advantages of sexual reproduction:
  - New gene combinations are created and so, by selection, some individuals may survive in the face of sudden environmental change.
  - Harmful mutations may be removed from the population.
  - Offspring may differ in their requirements due to the variation in the population and so there may be less competition for the same resources among offspring.
- Disadvantages of sexual reproduction:
  - It is costly in terms of time, energy and bodily resources.
  - Two organisms are required.

CHECK YOUR UNDERSTANDING 2.1c

1. List the male reproductive parts of a flower and describe the function of each part.
2. Draw and label the female reproductive parts of a flower and describe the function of each part.
3. Distinguish between pollination and fertilisation in plant reproduction.
4. Describe the pollination mechanism of two named Australian plants.
5. Identify two ways in which the reproductive structures differ between wind-pollinated and insect-pollinated plants.
6. Name the structure that each of the following develops into after fertilisation: ovum, seed, ovary wall.
7. Identify two methods of seed dispersal in named Australian plants.

2.2 Asexual reproduction – only one parent

Asexual reproduction means *not sexual* reproduction. The name tells us that it does not involve gametes (sex cells). Only one parent is required and all the genetic material in the offspring is passed down from this single parent. The result is that offspring are genetically identical to the parent and to each other. There is no production or fusion of gametes, and no mixing of genetic information to introduce variation. This has advantages and disadvantages. In unicellular organisms, asexual reproduction is the main form of reproduction. In multicellular organisms, sexual reproduction is more common.

The advantage of asexual reproduction is that it enables organisms to reproduce quickly without having to find a mating partner. For plants, which are immobile, finding a mate is complicated. Being genetically identical may give organisms a competitive advantage if they live in an environment to which they are particularly well adapted. Asexual reproduction among plants is more common in harsh environments where organisms are so specific that there is little benefit in having variation within the population. In these habitats, when favourable conditions arise suddenly, organisms can reproduce quickly and effectively.

Selective pressures that make asexual reproduction more effective than sexual reproduction include:

- a shortage of food and/or other resources – asexual reproduction uses less energy to produce offspring
- a small mating population and/or time and other constraints on finding a mate – only one parent is required for asexual reproduction.

The main disadvantage of asexual reproduction is that, with little or no variation in the population, the whole group (or species) is particularly vulnerable to sudden changes in the environment, such as drought, disease, or a new parasite or predator. Changes such as these may result in the survival of few, if any, individuals.

Many organisms, including protists, fungi and plants, alternate between sexual and asexual reproduction as a normal part of their life cycle. This mechanism, known as an **alternation of generations**, involves a sexually reproducing, gamete-bearing generation alternating with an asexually reproducing, spore-bearing generation.

Weblink
Alternation of generations in plants

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Asexual reproduction in plants

Have you ever planted the cut-off tops of carrots, beetroot or strawberries and discovered that they produce roots and grow into new plants? Or perhaps you have seen the runners that grow out of the ground in strawberry patches or around raspberry bushes. These are examples of asexual reproduction. New individuals arise from portions of the roots, stems, leaves or buds of adult individuals and are genetically identical to their parent. This type of asexual reproductive process is referred to as **vegetative propagation**.

Vegetative propagation

Some adult plants produce vegetative organs, such as bulbs, tubers, rhizomes and suckers, from which new plants can arise. This is equivalent to cloning an adult plant, as the offspring are genetically identical to the parent. **Perennating organs** (from the word *perennial*, meaning returning year after year) are underground organs such as roots or stems that contain enough stored food to sustain the plant in a dormant state, from one season to the next. These organs allow plants to survive adverse conditions such as extreme cold in winter or drought in summer. Even though the parts of the plant above the ground may die, the remaining underground organs develop buds that begin to grow once favourable conditions return. Buds on deciduous trees are also considered to be organs of perennation.

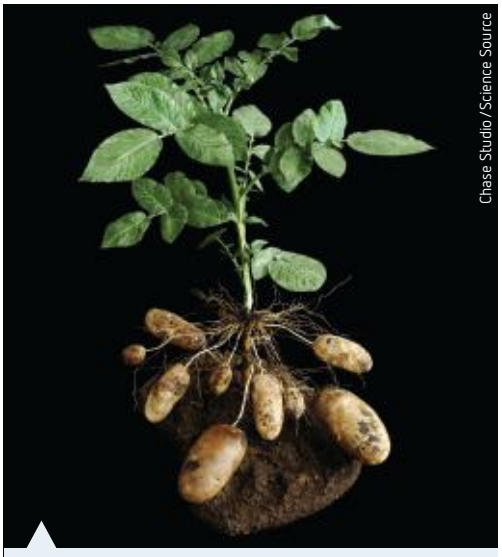

FIGURE 2.19 Potatoes develop tubers from swollen regions of the stem. New plants may also grow from the buds ('eyes') of a potato.

In addition to being a way of surviving from one year to the next, perennating organs, when separated, give rise to new plants and so they are a form of asexual reproduction. Gardeners often exploit this by splitting bulbs, cutting up rhizomes or runners, and taking cuttings of shoots from stems to grow new plants.

Vegetative propagation techniques are often used in agriculture – in growing perennial crops such as seedless grapes, watermelons and mangoes. This increases the production of crops when seeds are unavailable and/or difficult to germinate. Asexual reproduction is also used if plants have specific desirable traits that farmers want to perpetuate in future generations. Some common naturally occurring examples of vegetative propagation are described below.

Runners – modified stems

Runners are long, thin, modified stems that grow along the surface of the soil. In the cultivated strawberry, for example, leaves, flowers and roots are produced at every alternate node on the stem runner. Just beyond each second node, the tip of the node turns up and thickens, producing new roots and a new shoot that continues the runner. Another example is spinifex (Fig. 2.20), a grass that has long stems that grow horizontally along the surface of the soil. At each node, leaves and roots are produced and so the runner can be subdivided into new plants. Spinifex ensures its survival by sending out runners in harsh dune conditions such as high wind erosion, high salinity and high temperatures.

FIGURE 2.20 Spinifex is a type of grass (*Spinifex hirsutus*) with surface stems (runners) producing new leaves and roots (plantlets) at each node.

It is important to identify the organ that has the modification. For example, the runner is a modified stem, and the sucker, on page 50, is a modified root.


postolit/Shutterstock.com

FIGURE 2.21 New fern plants can grow when the rhizome (stem) is fragmented at the nodes.

Rhizomes – modified stems

Underground horizontal modified stems, or **rhizomes**, are characteristic of ginger, ferns such as bracken fern, and many grasses. They can give rise to a new shoot at each node. Gardeners often propagate ferns by splitting the rhizomes (Fig. 2.21).

Suckers – modified roots

The roots of some plants produce modified roots called **suckers** or sprouts, which give rise to new plants. Trees and shrubs that sucker, such as reeds, wattles and blackberries, can spread quickly into a vacant patch of habitat after disturbance. The colony wattle (*Acacia murrayana*) (Fig. 2.22) sends up shoots from the outer roots and these grow into separate plants if the parent shrub dies. This allows for rapid regrowth after a decline in numbers (after a bushfire or a drought, for example).

Apomixis

Some plants are able to produce offspring from special generative tissues, without involving fertilisation or the production of seeds, a form of reproduction known as **apomixis**. This 'generative tissue' may be in the form of gametes, such as in unfertilised ovules, or non-reproductive tissue such as leaf tissue (Fig. 2.23). It is termed generative tissue because it gives rise to plantlets that can produce asexual seeds. Apomixis is seen in plants such as kangaroo grass (*Themeda triandra*), lemon and orange trees (*Citrus*) and dandelions. Plantlets such as those that arise on leaves (Fig. 2.23) and their seeds grow into individuals that are genetically identical to their parent. The advantages of apomixis are that multiplication is rapid and the plantlets are able to produce seeds, increasing seed dispersal, an adaptation usually associated only with sexual reproduction. The disadvantage is the lack of variation that is typical of reproduction that involves two parents.

Apomixis also includes **parthenogenesis** in animals, a type of reproduction in which the new individual develops from an unfertilised egg produced by a female. You will have the opportunity to research this further in Investigation 2.2.


Infinity / Alamy Stock Photo

FIGURE 2.22 The colony wattle (*Acacia murrayana*) regenerates after drought or bushfire.


TIPAKORN MAKORNSEN/Shutterstock.com

FIGURE 2.23 Asexual reproduction by apomixis: *Kalanchoe* plantlets budding along the leaf margins

TABLE 2.3 Examples of the advantages of asexual reproduction

EXAMPLE	ASEXUAL REPRODUCTION MECHANISM	WHY IT IS AN ADVANTAGE
Spinifex grass	Stem runners put out leaves and roots at nodes along the ground	Enables reproduction in harsh conditions, requires less energy to reproduce by runner, and is very rapid
Colony wattle	Shoots grow from outer roots (suckers) and develop into separate plants	Rapid, and large numbers can be reproduced quickly, an advantage when rapid recovery is needed after a decline in numbers (e.g. fire or drought)

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INVESTIGATION 2.1

A practical and secondary-source investigation to examine organs of perennation in plants

Literacy

You are required to:

- examine organs of perennation in plants growing in your local area and/or in your garden at home
- conduct secondary-source research using a variety of sources to identify different types of perennating organs and how the modifications are adaptations to promote the continuity of the species
- present your findings in the form of a poster.

The poster you create will detail *two* examples of perennating organs that allow asexual reproduction in plants. The examples you select must include no more than one of each organ – root, stem or leaf. You may not use the organ outlined in the worked example below.

In the description on your poster, include:

- type of organ
- a scientifically named plant in which the organ occurs
- a labelled diagram of the organ, identifying plant tissues that:
  - store food
  - contain buds or other structures that give rise to new plants.
- a table in which you:
  - explain how the organ of perennation gives rise to new plants
  - discuss the advantages and disadvantages of the organ of perennation in terms of the survival of the species.

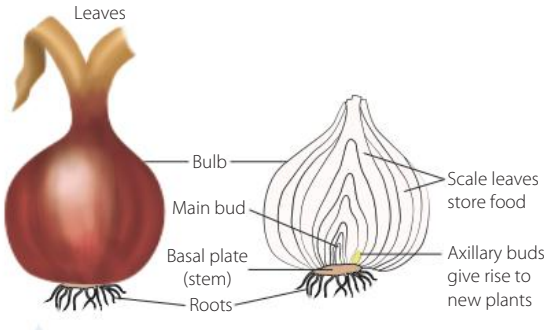
Some organs of perennation that you may wish to consider are:

- bulbs, corms and tubers
- epicormic buds
- organs of apomixis.
- rhizomes
- runners and suckers

WORKED EXAMPLE 2.1

ANNOTATED DIAGRAM AND DESCRIPTION OF AN ORGAN OF PERENNATION

Annotate a diagram of a perennating organ and explain the advantage of the organ of perennation to the survival of the species.

ANSWER	LOGIC
 Plant: *Allium cepa* **FIGURE 2.24** Stem bulb: onion (organ of perennation)	▪ Draw or insert a diagram. ▪ Label all parts of the organ of perennation. ▪ Annotate the part that stores food. ▪ Annotate the part that contains buds. ▪ Name the plant and the organ of perennation.

ANSWER	LOGIC
An onion is a stem bulb with fleshy leaves. The stem is reduced to a short disc, the basal plate. Roots grow on the lower surface of the disc and leaf bases on the upper surface.	▪ Describe the organ of perennation
Axillary buds occur at nodes where the leaves attach, and these buds can develop into new bulbs.	▪ Describe where the buds are.
If an onion bulb is planted, within a year or two there will be several bulbs in that place. These can be separated and re-planted to produce new plants.	▪ Explain the advantage of the organ of perennation to the survival of the species.

Scientists often use yeast for genome studies, because yeast is a simple eukaryote with similar cellular organisation and processes to those of complex multicellular organisms such as humans. The yeast genome was the first to be sequenced (in 1996).

Asexual reproduction in other organisms

It is not only members of the plant kingdom that undergo asexual reproduction. This type of reproduction is common in other organisms where it is advantageous for the parent and young to be genetically identical. Mechanisms of asexual reproduction in animals include budding, binary fission, sporogenesis, fragmentation and parthenogenesis.

Budding

In reproductive budding, an adult organism gives rise to a small bud, which separates from the parent and grows into a new individual.

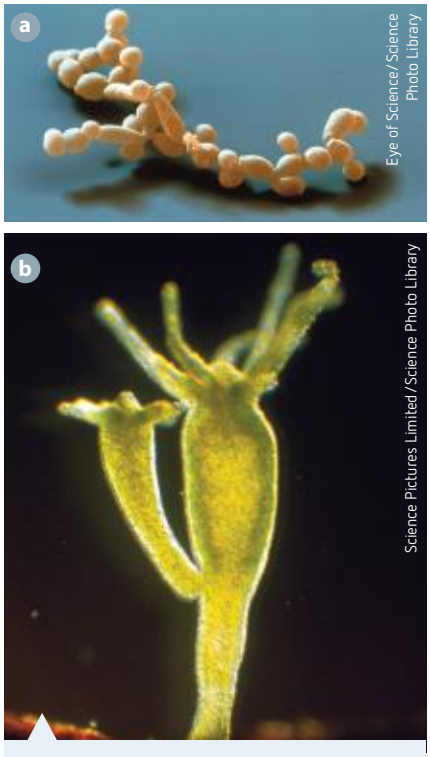


FIGURE 2.25 **a** Budding in yeast, where daughter cells remain attached to the parent cell; **b** hydra budding by producing a multicellular outgrowth from the side of the parent's body

Yeast are microscopic unicellular organisms that are classified as fungi, along with macroscopic moulds and mushrooms. There are hundreds of species of yeast; those with which you may be familiar are baker's and brewer's yeast (different strains of the same species, *Saccharomyces cerevisiae*) and the less useful *Candida albicans*, which causes digestive disorders and thrush in humans.

The cells of baker's yeast are oval-shaped and, when environmental conditions are favourable, a small outgrowth or bud develops on the parent cell. As this outgrowth enlarges, the parent cell replicates its DNA, the nucleus then divides and one copy moves into the bud or daughter cell. When the daughter cell reaches a certain size, it detaches from the parent cell and continues to grow, until it buds in turn. This continues as long as there are sufficient nutrients in the environment. Sometimes the buds do not break away from the parent cell and a chain of cells forms (Fig. 2.25a). The cell cycle of division in yeast is similar to that of human cells (Chapter 3). Yeast cells can replicate every 90 minutes and their numbers increase exponentially.

Yeast cells are also capable of sexual reproduction, but this is only triggered if there is a lack of nutrients. Yeast cells are unusual because they can exist in the haploid or diploid form, and both types of cells can reproduce asexually by budding. This makes them extremely useful in genetic studies. The diploid form is more resistant to harsh environmental

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conditions and can form spores that germinate when favourable conditions return, creating haploid yeast cells once again.

Budding also occurs in some multicellular organisms, including jellyfish, hydra (Fig. 2.25b) and grooved brain coral (Fig. 2.26). When conditions are favourable, the cells of the parent divide by mitosis and grow into a multicellular outgrowth, which develops into a smaller but identical individual or *bud*. This bud detaches from the parent and grows into a reproductive adult. One advantage of budding is that, if there is no variation in the environment, the identical offspring will always be adapted to their surroundings and survive to reproduce successfully. However, if the environment changes (for example, if a new disease or pest enters), the entire species may rapidly decline and die out.

Weblink
Yeast: an experimental organism for 21st century biology

FIGURE 2.26 Grooved brain coral

Binary fission

The term **binary fission** means splitting (fission) into two (binary). This is the main method of asexual reproduction in unicellular organisms such as bacteria (prokaryotes) and protists (unicellular eukaryotes). A newly divided cell grows to twice its size, replicates its genetic material (DNA) and then splits into two cells with identical genetic material (Fig. 2.27). This may seem simple, but for successful reproduction and survival, the timing of division is crucial and each individual must retain a complete and exact copy of the genetic material.

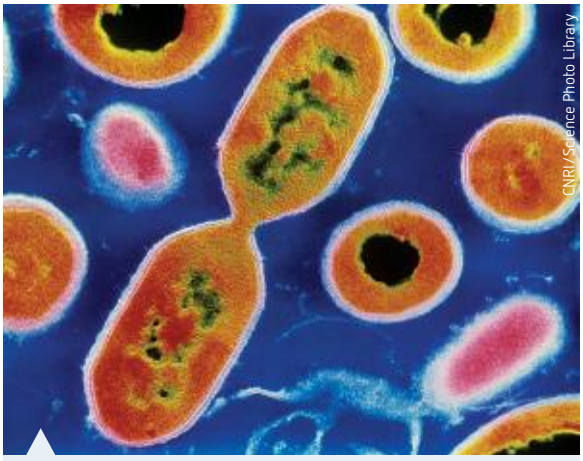

FIGURE 2.27 Transmission electron micrograph of the bacterium *Listeria* dividing into two by binary fission (×9800)

Binary fission in bacteria

When reproducing asexually, a bacterial cell can double in number every twenty minutes in favourable environmental conditions, thereby ensuring a rapid increase in numbers.

The prokaryotic cell grows to full adult size, then replicates its single DNA molecule and each copy of the DNA attaches to opposite ends of the cell membrane. A range of proteins accumulate at the centre of the cell and play a role in pinching off the cytoplasm and in ensuring that the DNA is not damaged in the process. A new cell wall is then synthesised in the area of cell **cleavage**. The new cells grow to full adult size before they divide again. The timing and sequence of the steps are closely controlled in the cell cycle.

Researchers are interested in finding out more about the regulation of binary fission in bacteria, as it may help them develop synthetic chemicals or find new antibiotics that interfere with the process of binary fission in pathogenic (disease-producing) bacteria, leading to new ways to combat bacterial infections.

A few bacterial species use other patterns of cell division to reproduce. Some bacteria grow much larger than the adult size and undergo multiple divisions, producing many offspring from one original cell, while others reproduce by budding.

Weblink
Reproduction in bacteria

Binary fission in protists

Mitosis and spindle formation are dealt with in more detail in Chapter 3.

Protists are unicellular eukaryotes that reproduce asexually by a type of binary fission (Table 2.5). This involves mitosis and the formation of a spindle within the cytoplasm of the cell to distribute chromosomes equally.

Amoeba is a single-celled organism that moves through water or damp soil by cytoplasm flowing out into long extensions of the cell called pseudopodia. It reproduces asexually by binary fission but, in adverse environmental conditions, it may form a cyst and then divide by multiple fissions inside the cyst, forming many identical cells that will be released when favourable conditions return. Binary fission in amoeba is termed 'irregular', as the cell is asymmetrical and division can occur along any plane (Fig. 2.28a).

TABLE 2.5 Types of binary fission in protists (Fig. 2.28 a–d)

TYPE OF BINARY FISSION	CLEAVAGE PLANE	EXAMPLE OF PROTIST
Longitudinal	Lengthwise	Flagellate forms, e.g. *Euglena*
Transverse	Crosswise	Ciliate forms, e.g. *Paramecium*
Oblique	At an oblique angle	*Ceratium*
Irregular	Along any plane	*Amoeba*

Weblink
Binary fission

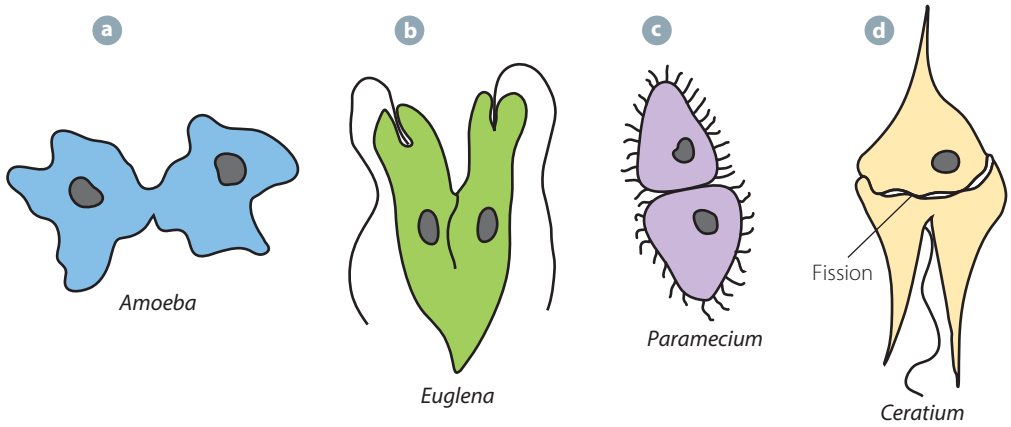

FIGURE 2.28 Binary fission in protists: **a** irregular cleavage in *Amoeba*; **b** longitudinal cleavage in *Euglena*; **c** transverse cleavage in *Paramecium*; **d** oblique cleavage in *Ceratium*

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Binary fission has the advantage of enabling rapid population growth over a short period of time in adverse conditions, as it requires only one parent. However, the main limitation is that no genetic diversity is generated.

Some protists are pathogenic and have a life cycle in two stages, one in an insect vector and one in a mammalian host. For example, in *Plasmodium* species that cause malaria, there are stages in both the vector and the host that are able to reproduce asexually by fission. The parasite reproduces sexually in the mosquito, and both sexual and asexual reproduction occur in a human host.

Pathogenic protists that belong to the group (phylum) protozoa and their life cycles are dealt with in more detail in Chapter 12, pages 419–20.

The Kingdom Protista is divided into phyla that include protozoa, slime moulds and algae.

Spores

Some organisms rely on **spore** production to reproduce. Spores are tiny, unicellular reproductive cells that are produced in great numbers by organisms such as fungi (moulds and mushrooms, for example) and some plants (mosses and ferns). Structures called **sporangia** (Fig. 2.29) produce very large numbers of spores, which are light and easily dispersed, travelling long distances by wind. Spores effectively expand the distribution of the species and are able to colonise new environments. Unlike a gamete, a spore does not need to fuse with another cell to produce a new individual. Spores also differ from seeds, as each spore is a single cell and therefore does not contain an embryo or a food supply. In terms of evolution, evidence shows that seed-bearing plants arose later than spore-bearing fungi and plants.

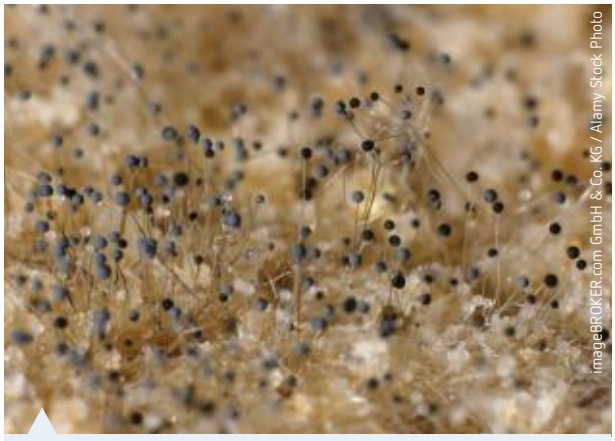

FIGURE 2.29 Bread mould, showing hyphae (white) and sporangia (black)

Multicellular fungi such as mould and mushrooms are made up of threads or filaments called **hyphae** (Fig. 2.29) that are branched and interconnected, forming the main fungal body, called the **mycelium** (plural *mycelia*). These threads may grow underground, or in dead and decaying matter. Unlike plants, fungi lack chlorophyll and do not photosynthesise, so they can grow in the dark. Their type of nutrition – either saprophytic (obtaining organic nutrients from dead and decaying matter) or parasitic (obtaining organic nutrients from living hosts) – allows fungi to colonise aerobic damp, dark environments where there is less competition for nutrients and water.

An example of spore formation in a fungus occurs in the black mould *Rhizopus nigricans*, which is similar to the pin-head mould *Mucor* and the antibiotic-producing mould *Penicillium*. These fungi are multinucleate, because there are many nuclei in the hyphae, with no cross walls to separate them into individual cells.

When environmental conditions are favourable, fungi reproduce asexually (Fig. 2.30). They develop large numbers of spore-producing units or sporangia, which grow upwards and are visible as the grey-green part of mould seen on bread, fruit and sometimes even leather. Sporangia develop as specialised tips of hyphal threads. They have numerous haploid nuclei, which develop into microscopic spores that are white at first and then turn black as they ripen. Each spore has several nuclei and some cytoplasm, surrounded by a wall. Spores are produced in enormous numbers and are extremely light, enabling widespread air dispersal. They carry genetic material identical to that of the parent. Under favourable environmental conditions, fungal spores germinate, absorbing water through the wall, which activates the cytoplasm to grow. Nuclear divisions occur, more cytoplasm is produced and the spore grows into a new mycelium. This process enables fungi to reproduce rapidly, colonise a wide area and ensure the continuity of the species.

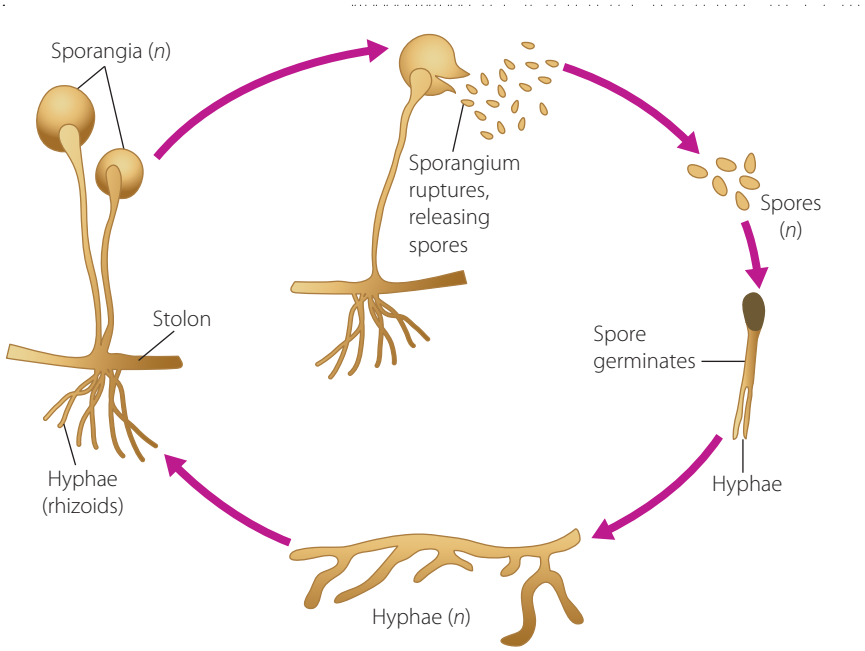

FIGURE 2.30 Asexual reproduction in a fungus such as *Rhizopus* (bread mould)

Weblink
Spores
Watch how sporangia release spores.

Weblink
Advantages of asexual reproduction in fungi

If a fungus is well adapted to a particular habitat, it is advantageous for it to conserve its genome and not introduce changes, such as those that would arise through sexual reproduction. Other advantages of asexual reproduction are that the fungus requires fewer nutrients and expends less energy. Retaining its adaptive advantage through asexual reproduction ensures the continuity of the species in its habitat.

However, changes in the environment will induce a sexual reproductive phase in fungi. During sexual reproduction, the hyphae of two different mating types fuse, before forming a sexual reproductive fruiting body that will have spores with different combinations of genetic material.

INVESTIGATION 2.2

Practical investigation of asexual reproduction (budding, binary fission, spore formation, propagation) in living organisms

Revise skills, risk assessment and safety for using a microscope from your Year 11 work.

INTRODUCTION

Most of the living organisms you will study in this practical undergo asexual reproduction under favourable environmental conditions. Simulating these conditions will make your investigation more successful. To expand your investigation skills, you may wish to design an experiment with conditions that you think will stimulate sexual reproduction in these organisms, and then test your hypothesis. You will need a microscope to examine asexual reproductive structures.

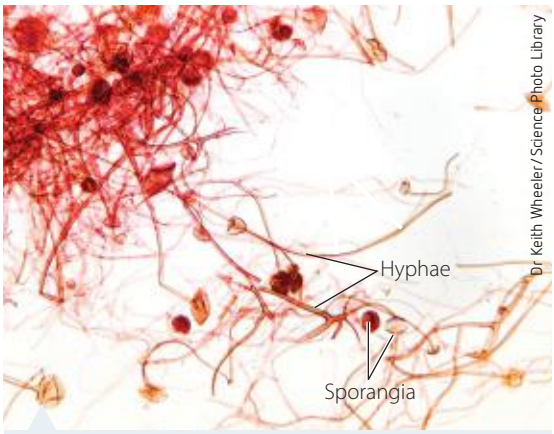

FIGURE 2.31 Sporangia at the end of hyphal threads in a *Mucor* species of fungus, as seen under the light microscope

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PART A

AIM

To investigate spore production in bread mould

MATERIALS

- 1 slice of moist bread (with no preservatives)
- 1 zip lock plastic bag or Petri dish
- dark environment such as a cupboard
- dissecting needle and forceps (1 per student group)
- glycerine in a dropper bottle (1 per student group)
- glass microscope slides and coverslips (1 per student group)

RISK ASSESSMENT

Copy and complete the risk assessment table below using your knowledge of microscope safety from your Year 11 practical work.

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?
Mould	Inhalation of spores may lead to allergic reaction	Wear a face mask when putting a fungal sample onto glass microscope slides. Use forceps and a probe; do not handle the mould. Wash hands at the end of the experiment.
Glass microscope slides	May break and cut you	
Microscope	Can drop on toes	
Electrical plug	Electrocution	

METHOD

1. Place a piece of damp bread (without preservatives) in a zip lock plastic bag or a Petri dish and keep in a warm, dark place for a few days. Damp oranges or cheese may also be used to grow mould.
2. Examine for mould growth with a hand lens daily. At first you may see hyphal threads (colourless at first). Once the mould has begun to darken, this signifies the production of spore bodies.
3. To prepare slides of bread mould (wear a face mask if you are allergic to spores):
   a. Place a drop of glycerine on a slide.
   b. Using forceps and a dissecting needle, place a few strands of hyphal threads from a pigmented area of the mould in the glycerine.
   c. Cover with a coverslip, lowering it carefully to avoid trapping air bubbles.
   d. Examine the slide under low and high power. Draw and label the mycelium and sporangia as seen under high power.

PART B

AIM

To investigate budding in yeast (unicellular fungus)

MATERIALS

- yeast (1 packet active dry baker's yeast)
- 80 mL warm water
- 15 g sugar
- slide and coverslip (1 per student group)
- dropper
- microscope (1 per student group)

METHOD

1. At least 2–3 hours before examining the yeast, combine the sugar, yeast and warm water, mixing well. (This step could be done the day before and the mixture left overnight.)
2. Place a drop of the yeast mixture on a slide, cover with a coverslip and observe under low and high power.

   Alternative method: If the yeast is on agar plates, pick yeast off the plate using a toothpick. Dip the tip of the toothpick containing yeast into a drop of water/methylene blue on a microscope slide.
3. Draw and label a diagram to show yeast budding, identifying the parent cell and the bud.

   Note: If methylene blue is used to stain cells, it is important to remember that yeast cells that are alive will appear opaque (enzymes in yeast actively break down the dye) and cells that appear blue are dead. Look for living yeast cells that are undergoing asexual reproduction by means of budding.
4. You may choose to film the process with a mobile phone through the eyepiece of the microscope.
5. If no active budding is seen, find a short video clip on the Internet, and watch the process of yeast budding.

PART C

AIM

To investigate budding and binary fission using prepared microscope slides

MATERIALS

Prepared microscope slides of:

- hydra budding
- amoeba undergoing binary fission
- yeast budding.

METHOD

1. Examine the prepared slides using a microscope, under low power and high power.
2. Identify the type of asexual reproduction observed in the named organism on the slide.
3. Draw and label the organism reproducing asexually. Write a heading for your diagram and record the magnification at which the specimen was viewed.

RESULTS

Copy and complete Table 2.6 to record the results of parts A to C of the investigation. Include accurately drawn scientific diagrams to show:

a. spore production in fungi
b. budding in yeast
c. binary fission in *Amoeba*
d. budding in *Hydra*.

Each row of your table will need to be large enough to include your diagram.

TABLE 2.6

NAME OF SPECIMEN	MODE OF ASEXUAL REPRODUCTION	DIAGRAM

CONCLUSION

Write a general statement summing up the forms of asexual reproduction that you observed in the variety of organisms examined in this investigation.

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PART D

AIM

To investigate other forms of asexual reproduction in plants and animals, using secondary sources and/or primary investigations

METHOD

Working in groups of 3–4 students, divide up the following tasks and carry out this part of the practical as a group.

1. Research a method and/or grow a plant using vegetative propagation – from, for example, a plant cutting, bulb, tuber or runner.
2. Find a video clip on the Internet *or* create a multimedia presentation that shows an example of the modes of asexual reproduction listed below. Create either a voice-over or subtitles to explain to your peers the mode of asexual reproduction and the advantages/disadvantages of this type of reproduction for the continuity of the species. You may conduct research and prepare a presentation on any organisms that display these forms of asexual reproduction – the organisms listed below are only examples.
   a. Fragmentation and regeneration – e.g. sea stars and flatworms, such as *Planaria*
   b. Parthenogenesis – e.g. Binoe's gecko (*Heteronotia binoei*), stick insects (Order *Phasmatodia*).

RESULTS

Present your results to your group. From the presentations, summarise in a table what you have learned from others in your group. Carry out peer assessment on the presentations created by each member of your group. Present the best one from your group to the whole class.

Some areas and questions you may wish to review when assessing the presentations of your peers are:

- Coverage – does the presentation cover the topic completely or partially, or is it too brief and just a summary?
- Illustrations – are they relevant and clear, and do they enhance understanding?
- Context – are the subtitles and/or voice-overs clear and relevant, and do they explain the advantages and disadvantages clearly and in context?

You may also wish to evaluate your team work. See the weblinks for ideas and scaffolds for peer assessment.

Personal and social capability

Weblink
Peer evaluation
Peer contribution – assessing the value of the contribution of members of our group

Weblink
Group evaluation
Assessing the effectiveness of your teamwork

DISCUSSION QUESTION

1. Copy and complete Table 2.7 to compare the processes of budding, binary fission and spore production in the organisms that you have reviewed in the presentations. Explain the conditions under which each occurs. Discuss the role of each mode of asexual reproduction in contributing to the continuity of species.

TABLE 2.7 Comparison of budding, binary fission and spore production

NAME OF SPECIMEN	MODE OF ASEXUAL REPRODUCTION	DIAGRAM	HOW IT ENSURES CONTINUITY OF THE SPECIES

EXTENSION

Asexual reproduction is not limited to protista, fungi and plants. Some animals are able to reproduce by means of budding and other forms of asexual reproduction. **Parthenogenesis** is a form of asexual reproduction in animals that falls into the category of apomixis (page 50). The term is derived from the Greek word '*Parthenos*' meaning virgin, and '*genesis*' meaning creation. Parthenogenesis involves the production of offspring from an unfertilised gamete, usually an egg (ovum). This unusual form of reproduction, although classified as asexual, may also be thought of as 'incomplete' sexual reproduction. It is common in some insects (aphids, ants, bees and wasps, for example), and has been found to occur fairly commonly in reptiles as well. The young that are produced are genetic clones of their mother. There are many Australian lineages that demonstrate parthenogenesis. (See the weblink.) This raises a good question for discussion: Is there a need for males in animal species that have parthenogenesis? Research the advantages and disadvantages of parthenogenesis and prepare arguments for and against the discussion question posed.

Weblink
Parthenogenesis

KEY CONCEPTS

- Budding occurs in yeast and some multicellular organisms such as *Hydra* and brain coral.
- Binary fission is the most common form of asexual reproduction in bacteria and protists.
- Asexual reproduction in bacteria occurs in a series of steps: a cell grows to twice its size; DNA replicates; DNA separates; protein accumulates at the cleavage site; the cytoplasm divides; and a new wall is synthesised.
- Both binary fission and budding involve only one parent, so no mate is required, allowing rapid proliferation of genetically identical organisms that are well suited to survive in their particular environment.
- Disadvantages of asexual forms of reproduction: lack of genetic variation in offspring, which may lead to their demise in unexpected harsh conditions or as a result of other changes in selecting agents; the large numbers of offspring produced may compete with each other for resources.

CHECK YOUR UNDERSTANDING 2.2

1. Using examples, explain how the transfer of gametes during sexual reproduction occurs in plants and animals.
2. Describe two pollination mechanisms in plants and explain how they ensure the continuity of the species.
3. Describe two reproductive mechanisms in named examples of animals, to ensure the survival of the young.
4. In a table, compare the structure of a gamete, a seed and a spore, and explain the role of each in reproduction, using examples.
5. In a table, compare forms of asexual reproduction including budding, binary fission and propagation.

2.3 Sexual reproduction in mammals

Worksheet
The male and female reproductive systems

As you now know, from reading section 2.1, sexual reproduction involves fertilisation – the fusion of male and female gametes (sex cells) to form a zygote (fertilised egg). The zygote contains a new combination of genetic material from both parents. Because a variety of offspring are produced, this genetic variation increases the chances of survival of the population in the case of environmental change. For the energy expenditure of sexual reproduction to be worthwhile, the chances of reproductive success must be maximised.

Mammals have several reproductive mechanisms to maximise reproductive success, including:

- *internal fertilisation* to increase the likelihood that gametes will meet (this occurs in all three subclasses of mammals)
- *implantation* of the embryo into the uterine wall, with internal development of the embryo (marsupials and eutherians) (Fig. 2.32) to increase the embryo's chance of survival
- *pregnancy* to allow the developing young to be protected from the external environment, have a constant nutrient supply and complete a gestation period (short in marsupials; prolonged in eutherians, whose young are well developed when they are born).

The three classes of mammals are:

- monotremes (egg-laying)
- marsupials (pouched)
- eutherians (placental).

All stages of sexual reproduction are carefully timed and synchronised by a combination of hormones that coordinate the reproductive cycle to ensure greater reproductive success.

Strategies such as the timing of reproductive cycles, embryonic development in utero, reduced number of young produced and quality of parental care have evolved to further increase the likelihood that the young will develop and grow to become reproductive adults. All stages of sexual reproduction in mammals, from gametogenesis to courtship behaviour, pregnancy and birth, are carefully regulated by hormones. Successful offspring will survive to become adults, competing in turn for mates of their own. The cycle must be self-perpetuating, to ensure the continuity of the species.

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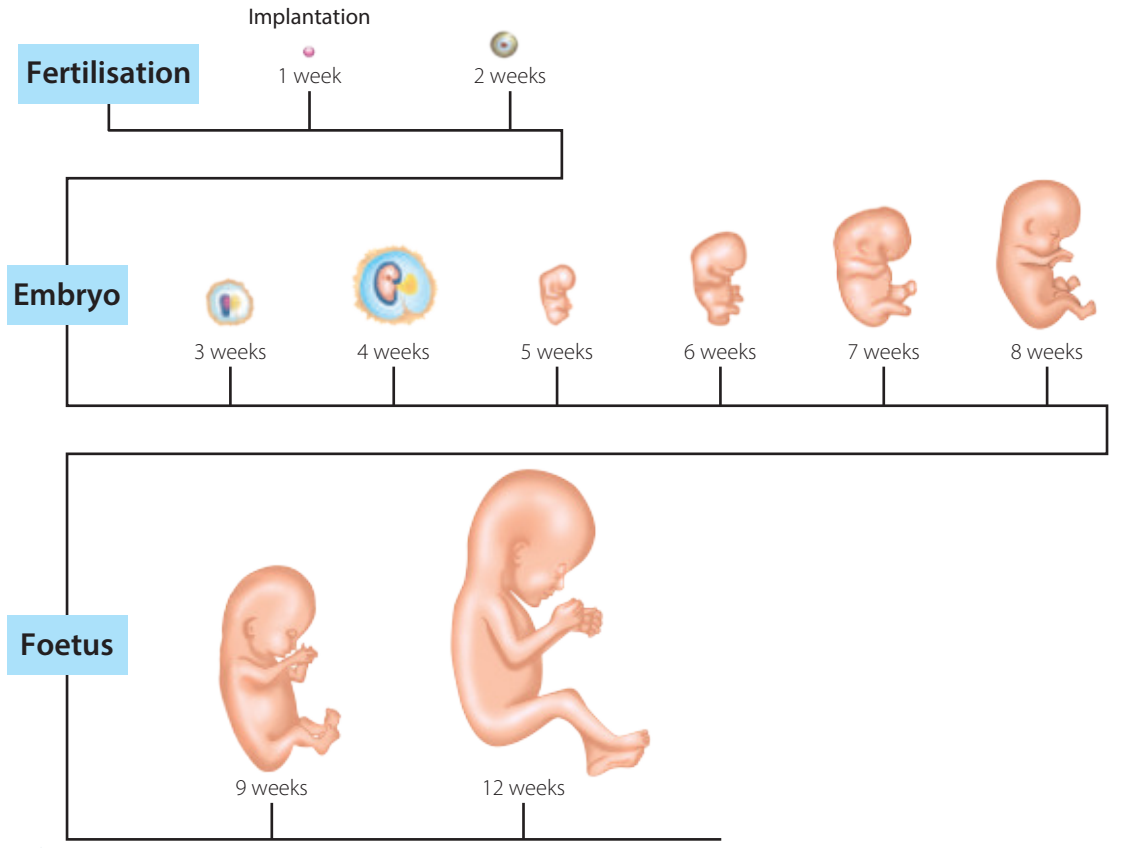

FIGURE 2.32 Fertilisation and embryonic development in humans, to foetus stage

Hormones

Hormones are chemical substances that act as messengers in the body, coordinating many aspects of functioning, including metabolism and reproduction, so that actions within the body are synchronised.

The **pituitary gland** is an endocrine gland about the size of a pea, attached to the base of the brain and just above the roof of the mouth. Referred to as the master gland, it secretes hormones that stimulate or inhibit other endocrine glands, regulating the release of their hormones for growth, metabolism and reproduction.

Hormones that specifically affect the growth or functioning of the reproductive organs or the development of secondary sex characteristics are called **sex hormones**. They are produced in special tissues in the ovaries and testes and in the pituitary gland and adrenal cortex. Hormones play a vital role in all aspects of the development and functioning of the male and female reproductive systems. The reproductive organs in mammals are present at birth, but only mature and begin their reproductive function when stimulated by hormones secreted during puberty.

In humans, puberty generally occurs between age ten and fourteen in girls, and twelve to sixteen in boys. The **gonads** (reproductive organs) become functional at puberty and the reproductive cycle commences. Gametes are produced in the male and female gonads by a process known as **gametogenesis**.

See Chapter 14, pages 488–92 for more information on hormones.

Hormonal control of breeding seasons

Hormones regulate the sexual behaviour of mammals by limiting the ability of some mammals to reproduce to certain times of the year, known as breeding seasons. Breeding cycles in these mammals involve periods of female fertility being limited to once or twice a year. In these **seasonal breeders** (such as sheep and cattle), mating occurs only during periods of female fertility, commonly referred to as the animal being 'on heat' or 'in season'. The biological term is that the animal is 'in **oestrus**'.

Weblink
Hormones in the oestrus cycle

Weblink
Detecting oestrus in a farm animal

Higher order primates (including humans) and some other mammals (such as pigs, rats, mice and rabbits) are called **continuous breeders**. Female fertility occurs in a cycle that repeats throughout the year and, rather than having a breeding season, these animals are sexually active all year round.

There are advantages to both types of breeding cycle. The young of mammals that have a limited breeding season are usually born when temperatures are warm and food is plentiful, increasing the chance of offspring surviving. The parents benefit from the reduced period of time during which mating, gestation and raising of young takes place, a period during which their energy reserves become limited and their vulnerability to predators increases. The obvious advantage to continuous breeders is the ability to reproduce all year around.

In the discussion that follows, humans are used as an example to explain the reproductive cycle of mammals. The oestrus cycle in seasonal breeders is similar in physiology and hormonal control, the main difference being in the timing.

Hormones involved in mammalian reproduction

There are three types of sex hormones.

- **Androgens** (from the Greek *andro* meaning 'male') are commonly referred to as male hormones. Androgens control the development and functioning of male sex organs and **secondary sex characteristics** such as deepening of the male voice, increase in the growth and thickness of hair and in the size of muscles and bones. Cells in the testes secrete the androgen **testosterone**, which plays a primary role in **spermatogenesis** (production of sperm).

  Androgens are present in both males and females, and their production increases during puberty in both sexes, but their levels are much higher in males. Androgens are precursors (margin note) of oestrogens (a group of female hormones).

A precursor molecule is a chemical that is transformed into another compound.

- **Oestrogens** (from the Greek word *oistros* meaning 'mad desire', referring to willingness of a female to mate when she is at the fertile part of her cycle) are the main group of female hormones. They control the development and functioning of the female reproductive system and secondary sex characteristics, such as enlarged breasts, pubic hair and widening of the hips.

  Oestrogens are present in both males and females, but occur in much higher levels in females of reproductive age. In males, together with testosterone, oestrogens play a role in the maturation of sperm. Oestrogens are responsible for the onset of oestrus (in heat) just before ovulation in female mammals that are seasonal breeders. The main function of oestrogens in mammalian reproduction is *ovarian functioning* and therefore *fertility* in females.
- **Progestogens**, a second group of female hormones, have been named from the words *pro* (one) and *gestare* (latin for 'to carry about', as in *gestation*). **Progesterone** is the most common progestogen and it plays a primary role in *pregnancy*. It also stimulates the secretion of milk in mammary glands (*lactation*) and a drop in its levels plays a role in initiating menstruation.

  Using current biotechnology, female hormones can be artificially manufactured and used either as a form of contraception to prevent pregnancy, or to increase fertility and help women become pregnant. For example, **progestin** is the synthetically produced version of the hormone progesterone. Oestrogens and progesterone may be used in combination or individually. Oestrogens and progestogens may also be used in:
  - hormone replacement therapy for women during menopause (if required)
  - the treatment of prostate cancer and some forms of breast and endometrial cancers.

Hormonal control of the female reproductive cycle

Endocrine glands regulate and control the ovarian and menstrual cycles in a coordinated manner (Fig. 2.33), synchronising these cycles to ensure fertility. The result is an increase in the probability of successful reproduction, biological fitness and, therefore, the continuity of the species.

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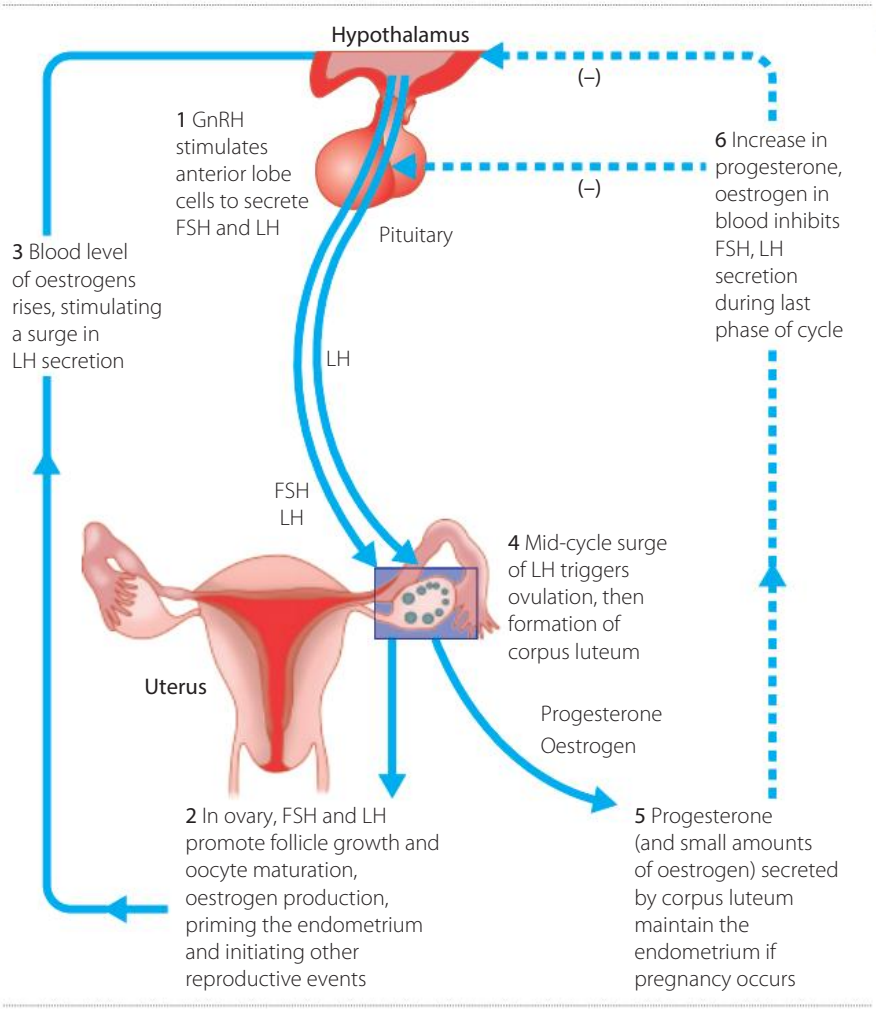

FIGURE 2.33
Hormone production and action in the female reproductive system

FHS = follicle-stimulating hormone

LH = luteinising hormone

GnRH = gonadotrophic-releasing hormone

In humans, the **pituitary gland** secretes a number of hormones that regulate other endocrine glands, including the ovaries of females. Hormones of the pituitary and gonads are therefore the key players in regulating reproductive cycles in mammals.

Oestrogen and progesterone, produced by the ovaries and controlled by hormones of the pituitary, regulate the:

- **ovarian cycle** by controlling the production and maturation of gametes (ova) in the ovaries
- **menstrual cycle** by preparing the uterus for implantation of a fertilised egg each cycle; if fertilisation does not take place, the levels of oestrogen and progesterone decrease and, as a result, the lining of the uterus tears away, accompanied by bleeding, known as **menstruation**
- maintenance of pregnancy
- preparation for and maintenance of lactation.

The pituitary secretes two **gonadotropic** hormones:

- **follicle stimulating hormone (FSH)**, which is important for stimulating maturation of follicles in the ovaries of females
- **luteinising hormone (LH)**, which promotes final maturation of the ovarian follicle, ovulation and development of the corpus luteum in females. It also stimulates the secretion of testosterone.

In addition to gonadotropic hormones, the pituitary gland secretes a **lactogenic hormone** called **prolactin** in females. This acts on breast tissue to prepare for and maintain milk production to suckle the young.

The interaction of the pituitary gland with the ovaries and testes is synchronised by feedback loops, which you will learn more about in Chapter 14.

Weblink
Revise female reproductive hormones and their functions

The balance in the levels of sex hormones in the body at any point in time determines the fertility of a female and whether she can conceive a child.

The ovarian cycle

Female babies are born with all the eggs (ova) that they will produce during their lifetime, but these ova are immature and have partly developed in their ovaries. During puberty, the ovarian and menstrual cycles begin (referred to as **menarche**). The ova in the ovaries become surrounded by a single layer of cells that envelop them and begin to divide, resulting in the formation of primary (dormant) follicles in the ovary (Fig 2.34). Hormones that are secreted during puberty (and for approximately the next 36–40 years) trigger the development and maturation of ova each month (except during periods of pregnancy), until **menopause** is reached. At the start of each monthly cycle, a few follicles begin to develop, but usually only one enlarges more than the others to reach maturity (Fig. 2.34). In humans, the cycle repeats approximately every 28 days, although this varies from one female to another.

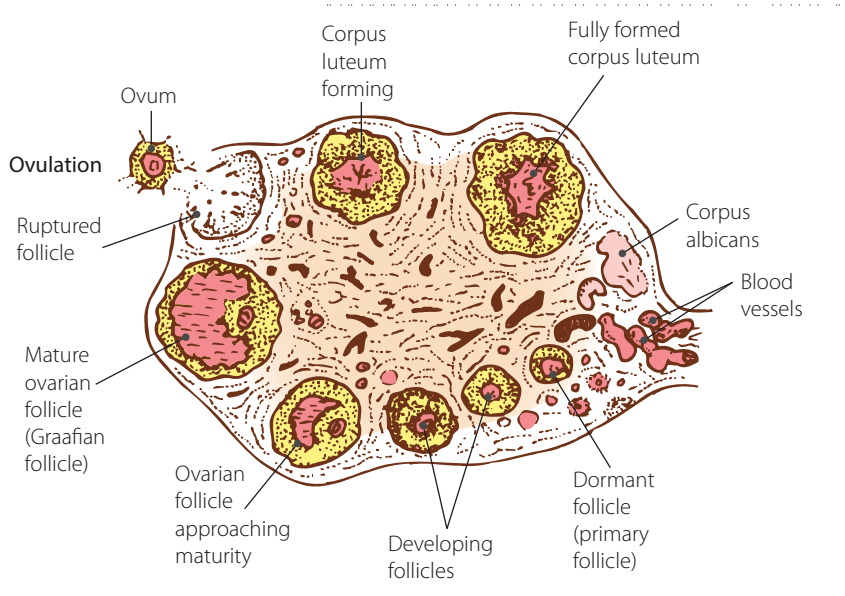

FIGURE 2.34 Diagrammatic representation of the ovarian cycle, including ovulation

The follicular phase

The follicle cells secrete fluid, which pushes the egg to one side of the follicle. The enlarged, dominant follicle moves to the surface of the ovary and creates a bulge. It is now mature and termed a **Graafian** follicle. The development from primary follicle to Graafian follicle takes approximately 10–14 days.

During this follicular phase, the cells lining the follicle secrete **oestradiol** (an oestrogen hormone). This causes a surge in the production of LH, which results in ovulation. The LH surge also stimulates the next phase of the ovarian cycle, during which the corpus luteum forms and progesterone is synthesised.

The Graafian follicle bursts, releasing the egg – a process known as **ovulation**. The funnel-shaped, open ends of the uterine tubes contain cilia that beat, drawing the egg into the tube. The egg (with some follicle cells still clinging to it) moves down the tube towards the uterus. If sperm are present, fertilisation may occur (page 67). Ovulation usually occurs mid-cycle (Table 2.6, page 66). Once released, the female ovum is only viable for 12–24 hours.

The luteinising phase

The luteinising phase is usually 14 days. It begins after ovulation, when the burst follicle in the ovary enlarges and changes colour, building up a yellow protein called lutein. The large mass of vacuolated cells is now called the **corpus luteum** (Latin for 'yellow body'). The corpus luteum secretes the hormone **progesterone**, which acts on the uterus, preparing it for pregnancy.

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The menstrual cycle

The cycle of changes in the ovaries is accompanied by a cycle of changes in the uterus, known as the menstrual cycle (Fig. 2.35). An average menstrual cycle repeats every 28 days, but there can be considerable variation in the actual length of the cycle.

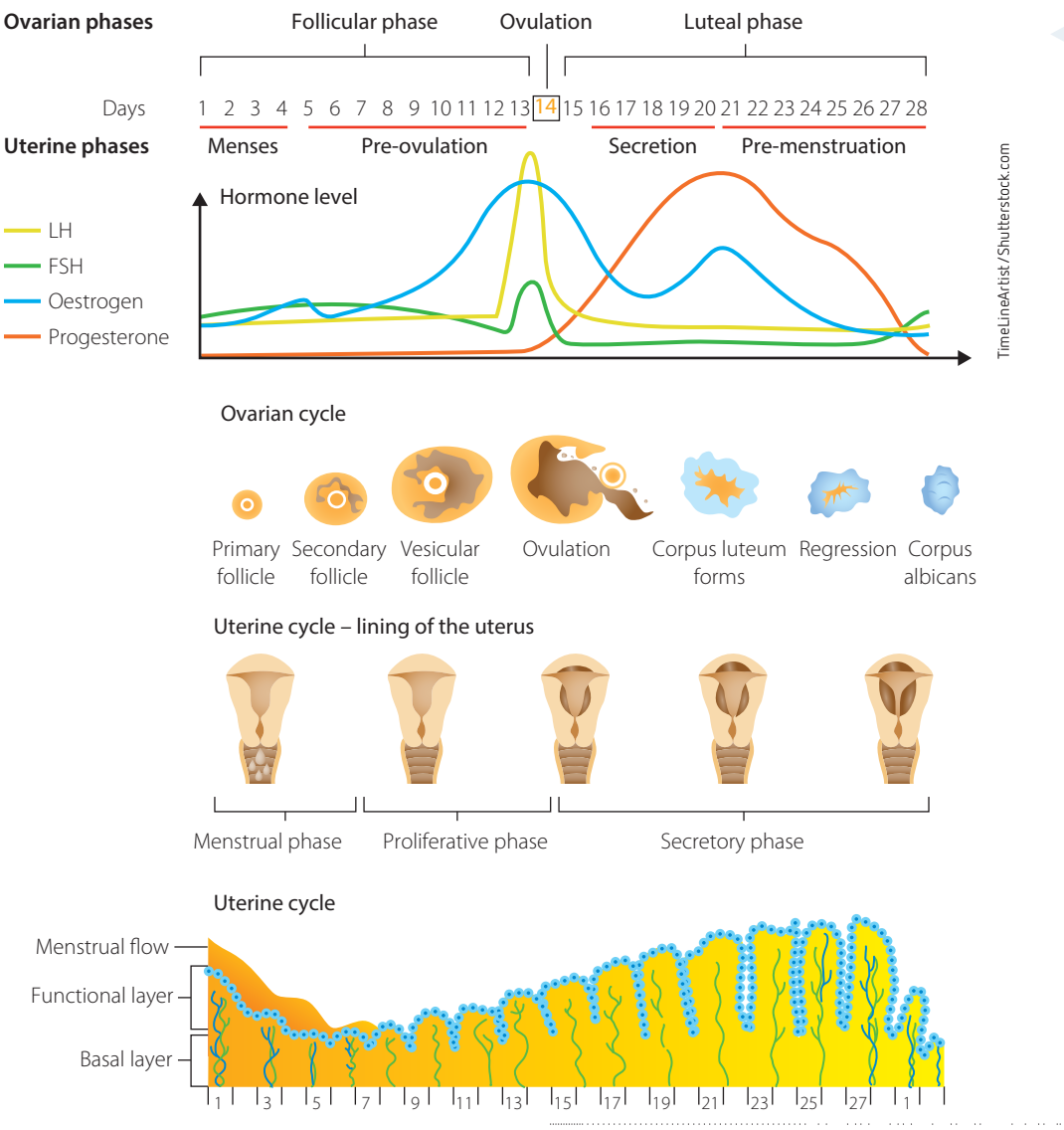

FIGURE 2.35 Hormonal control of the ovarian and menstrual cycles

The menstrual cycle starts with **menses** (the menstrual period), which lasts about four days. During this time, the **endometrium** (lining of the uterus) breaks down and tears away. This is accompanied by bleeding, which is known as menstruation. The first day of menses marks the beginning of the follicular phase, which ends on the day of ovulation (Table 2.6).

Following menstruation, a new endometrial lining forms in the uterus over about 5–12 days, known as the pre-ovulation phase. Ovulation takes place in an ovary about 13–15 days after the start of menstruation, but this timing varies from person to person.

After ovulation, the corpus luteum, which is enlarging within the ovary, secretes the hormone progesterone, as well as some oestrogen, into the bloodstream. Progesterone acts on target cells in the uterus, preparing the endometrium for implantation of the fertilised ovum and pregnancy. The endometrium becomes highly vascularised, reaching a peak eight or nine days after ovulation (secretory phase), around the time of expected implantation. The endometrium has glands that secrete a watery mucus in this phase.

Weblink
Ovarian and menstrual cycle

If a fertilised ovum implants in the uterus, pregnancy results and the uterine wall is maintained by the secretion of progesterone and oestrogen. These are produced by the corpus luteum at first and later by the placenta. During pregnancy, a placenta forms, attaching the developing embryo to the uterine wall. The placenta secretes hormones including progesterone, oestrogen and human chorionic gonadotropin (HCG) to maintain pregnancy. Once the placenta is able to secrete these hormones, the corpus luteum in the ovary begins to degenerate.

Weblink
Hormonal control of the menstrual cycle

TABLE 2.6 Stages in the ovarian and menstrual cycles

STAGE	TIME SPAN* (DAYS)	EVENTS
Menstruation	1–4	Uterine bleeding, accompanied by shedding of the endometrium
Pre-ovulation	5–12	Endometrial repair begins; development of ovarian follicle; uterine lining gradually thickens
Ovulation	13–15	Rupture of mature follicle, releasing egg
Secretion	16–20	Secretion of watery mucus by glands of endometrium, cervix and uterine tubes; movement and breakdown of unfertilised egg; development of corpus luteum
Pre-menstruation	21–28	Degeneration of corpus luteum; deterioration of endometrium

*Time spans are average, but vary widely between women.

Hormonal control of the male reproductive cycle

The production of sperm, or spermatogenesis, is controlled by hormones. In humans, spermatogenesis involves the interaction of three glands: the hypothalamus in the brain, the pituitary gland at the base of the brain and the **Leydig cells** in the testes. In males, LH stimulates the production of testosterone and FSH stimulates the production of a protein by Sertoli cells in the testes, to maintain testosterone at a level high enough to promote spermatogenesis. When the hormone inhibin is secreted, it reduces the levels of FSH in the body (Fig. 2.36).

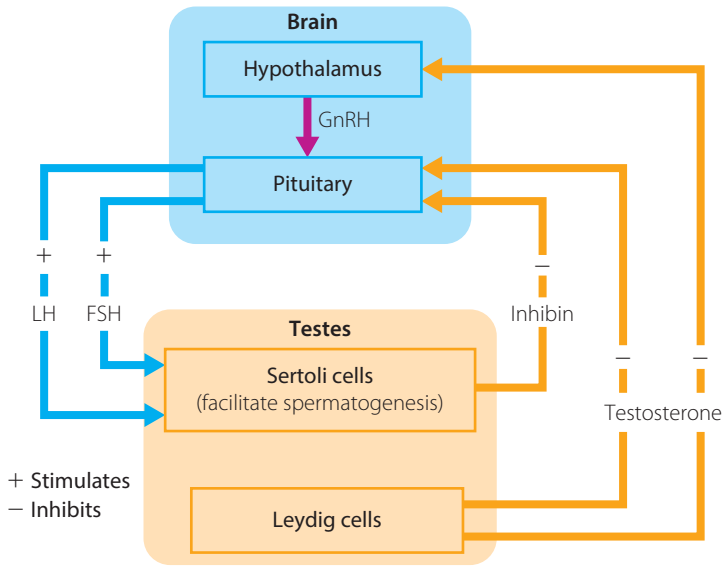

FIGURE 2.36 Hormonal control of sperm production: LH and FSH from the pituitary stimulate spermatogenesis and testosterone secretion by the testes. Testosterone inhibits the secretion of GnRH by the hypothalamus, and testosterone and inhibin from the testes inhibit the secretion of LH and FSH by the pituitary.

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Sperm production in the male

Sperm are produced by meiosis inside sperm tubules in the testes, and are stored until they are mature. Each sperm is microscopic and shaped like a tadpole, with an enlarged head that contains the haploid nucleus and a long tail that whips from side to side to enable the sperm to move. During copulation, semen containing sperm is introduced into the vagina of the female, and must undertake an epic race to reach the egg cell. Although half a million sperm may begin the journey, only a few hundred to a few thousand will reach the ovum. (See the image on page 33.) Contractions of the vaginal wall assist the movement of sperm. The whipping action of the tail of the sperm is thought to keep the sperm afloat in the semen and assist the sperm in penetrating the egg. Sperm can survive in the female reproductive tract for a couple of days.

Fertilisation

Sperm are attracted to the egg by **rheotaxis** – movement through a fluid – for internal fertilisation. Oviducts secrete a fluid that travels down the female reproductive tract, and sperm swim upstream (positive rheotaxis). Sperm that reach the oviduct are held in storage and are released in small batches. The presence of progesterone and an alkaline pH cause sperm to mature so that they can penetrate the egg. These sperm become hypermobile and their tails beat strongly to propel them towards the egg.

When sperm reach the egg cell, they must cross three layers. They physically push through the first membrane (Fig. 2.37), which still has follicle cells attached. These protective cells release enzymes to assist penetration by the sperm. When the acrosome (protective cap) of a sperm comes into contact with glycoproteins of the next barrier, the zona pellucida, the acrosome fuses with the cell membrane of the sperm head, allowing the tip of the sperm to release enzymes that assist its penetration. Many sperm may reach the cell membrane (plasma membrane), which is the last barrier. Surface proteins allow only one sperm to penetrate this barrier, triggering the release of enzymes by the egg that destroy the glycoproteins in the zona pellucida and cause electrical changes, preventing other sperm from entering. The first sperm to penetrate this inner barrier causes the ovum to immediately undergo its second meiotic division.

Worksheet
What are the social and ethical implications of applying IVF technology?

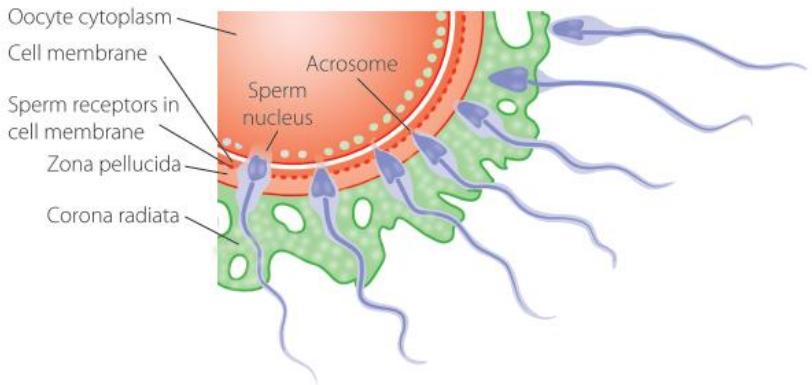

FIGURE 2.37
Acrosome reaction during fertilisation. A single sperm succeeds in burrowing through the corona radiata and zona pellucida and making contact with the oocyte's cell membrane. The sperm's cell membrane fuses with that of the oocyte and the sperm releases its nucleus into the cytoplasm of the oocyte.

Fertilisation occurs when the haploid nucleus of the egg fuses with that of the sperm, forming a diploid fertilised egg, called the **zygote**. At this stage, most epigenetic markers from the parents are 'wiped' off the DNA, so all genes are switched on – how this occurs is an area of current research. Following fertilisation, the egg divides as it travels along the oviduct and begins developing into an embryo.

See Chapter 4, pages 131–2 for information on epigenetic markers.

Weblink
Structure and function of the placenta and secretion of hormones

Hormonal control of pregnancy and birth

When the embryo implants into the uterine wall, this marks the beginning of pregnancy.

Once the embryo implants, the corpus luteum in the ovary continues to grow and secrete hormones for the first three months of pregnancy. In the latter six months of pregnancy, the corpus luteum shrinks and degenerates slowly (although it is still present in its degenerate form at childbirth). The placenta takes over the role of producing hormones to maintain pregnancy. Following pregnancy, the ovarian cycle usually only resumes once the mother stops breastfeeding the baby.

If the egg is not fertilised, the corpus luteum begins to degenerate about 8–10 days after ovulation, forming a mass of fibrous tissue known as the **corpus albicans**. An unfertilised egg degenerates during the phase known as *pre-menstruation*, after which menstruation occurs and the ovarian cycle begins again.

FIGURE 2.38 Baby developing in utero (note the umbilical cord and placenta)

The environment in which the baby develops (Fig. 2.38) is rigorously controlled by hormones and is important not only for the growth and development of the foetus to birth, but also for its long-term health.

Hormones that maintain pregnancy are secreted by the pituitary gland and ovaries of the mother at first, but once the placenta is established it begins secreting hormones too (Fig. 2.39).

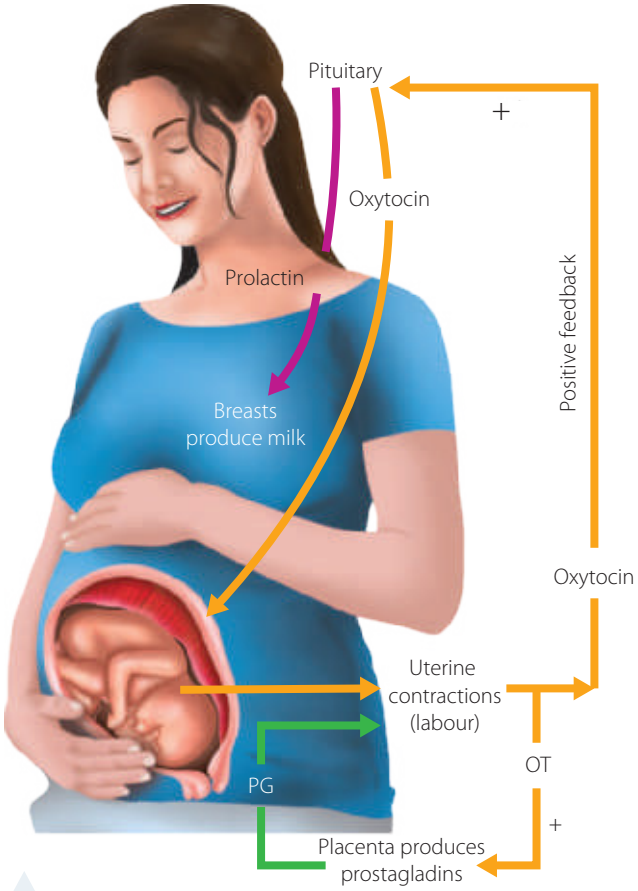

FIGURE 2.39 Hormone secretion during pregnancy and birth. OT: oxytocin, PG: prostaglandins

The levels of oestrogen and progesterone are optimised during the ovulation cycle to create ideal conditions for implantation. Oestrogen promotes the growth of the endometrium (lining) of the uterus and progesterone stimulates the secretion of mucus by the cells lining the endometrium and the growth of blood vessels. Once implantation has occurred, the main role of progesterone becomes that of suppressing uterine activity, thereby supporting foetal development and reducing the risk of the foetus being disturbed or expelled by uterine contractions. Progesterone also reduces the mother's immune response to foetal antigens. Adequate progesterone production by the corpus luteum is essential in maintaining the pregnancy until the placenta takes over this function at around seven to nine gestational weeks.

Other hormones are also important in maintaining pregnancy and ensuring the normal development and growth of the baby. Some hormones prevent foetal overgrowth and others inhibit overgrowth at various stages of pregnancy, mainly by regulating the amount and types of nutrients that cross the placenta. Table 2.7 outlines the hormones that coordinate foetal development and maintain pregnancy.

The placenta is a large disc of tissue formed of both maternal and foetal tissue (Fig 2.40). Each side produces finger-like extensions, called villi – extensions from the uterine wall of the mother form some villi, which interlace with extensions from the chorion membrane surrounding the foetus (chorionic villi). The placenta, embedded in the uterine wall, is connected to the foetus by an umbilical cord, which carries blood vessels to and from the baby. The role

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of the placenta is to transport oxygen and nutrients from mother to foetus and to remove wastes such as carbon dioxide and urea from the foetus and pass them to the mother for excretion. These gases, nutrients and wastes diffuse across the narrow space between the villi. The mother's and baby's blood do not come into direct contact.

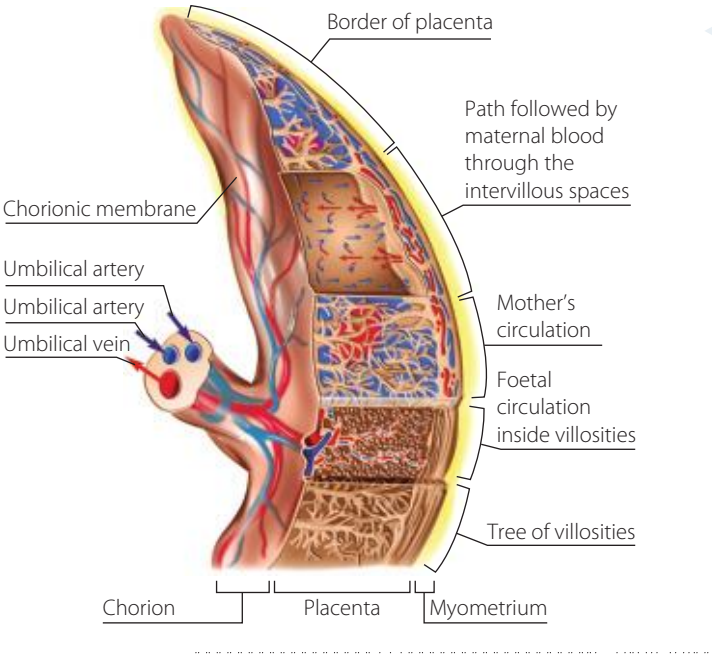

FIGURE 2.40 Placenta showing maternal and embryonic tissues and blood supply

TABLE 2.7 The role of hormones in pregnancy and foetal development

HORMONE	SECRETED BY	ACTIVE PERIOD DURING PREGNANCY	EFFECT ON PREGNANCY	EFFECT ON FOETAL DEVELOPMENT
Maternal thyroxin	Mother's thyroid gland	From 12 gestational weeks	Low levels increase the risk of detachment of the placenta and pregnancy loss	Low levels result in low birth weight and immature lungs
Insulin	Foetal pancreas (mother's insulin does not cross the placenta)	From 10 gestational weeks	No effect	Promotes foetal growth, allowing glucose to become available for tissue growth and fat deposition
Human chorionic gonadotropin (hCG)	Developing embryo	First trimester of pregnancy. Note: pregnancy tests measure the presence of this hormone to confirm a pregnancy	Maintains the corpus luteum, which in turn helps maintain the endometrium	No effect
Insulin-like growth factors (IGF)	Uterine wall of mother	No effect	IGF I: alters placental transport of nutrients, matching this to foetal demand IGF II: stimulates growth of the placenta	Promotes growth of foetal tissue
Glucocorticoids (cortisol and cortisone)	Foetus	Increase during third trimester Elevated if mother becomes stressed or anxious	Alcohol intake by the mother during pregnancy stimulates glucocorticoid secretion, which leads to low birth weight and affects organ development	Limits foetal growth; promotes and coordinates the specialisation and development of foetal tissues and organs (e.g. brain, heart and lungs)

Weblink
Hormonal regulation of labour and birth

Birth

Two things must happen for the baby to be born.

- The muscles in the uterus must contract to expel the baby.
- The tissue of the cervix must soften so that the cervix can dilate (widen) to allow the passage of the baby.

What triggers the process of childbirth is largely unknown. **Prostaglandins** secreted by the wall of the uterus initiate labour. They act by making the tissue in the uterus more sensitive to another hormone, **oxytocin**.

Oxytocin promotes coordinated contraction of the smooth muscle of the uterus and softening (ripening) of the cervix, so that the baby can be born. The hormone **relaxin** is also produced and this further aids the softening of the cervix.

Progesterone and oestrogen levels decline during labour and contractions become stronger.

As the contractions increase in intensity, beta-endorphin hormones are released. These act as a natural form of pain relief and also promote feelings of elation. Very close to the birth, a surge of adrenaline is released by the mother's body and this causes some very strong contractions that lead to the birth.

After the birth of the baby, oxytocin secretions continue for a while, causing the uterus to contract and expel the placenta (sometimes referred to as the 'afterbirth'). The contractions also limit blood flow to the uterus, reducing the risk of the mother bleeding.

Prolactin is a hormone produced mainly in the pituitary gland (but at times also in the uterus and breasts). It stimulates milk production in the breasts (Fig. 2.39). Levels of prolactin begin to rise during the second trimester of pregnancy and this causes enlargement of the mammary glands for milk production. Prolactin and oxytocin release are further stimulated by suckling of the baby. Prolactin and oxytocin are also thought to play an important role in maternal behaviour and bonding.

KEY CONCEPTS

- Sperm are attracted to the egg by (positive) rheotaxis – movement (upstream) through a fluid.
- Fertilisation occurs when the haploid nucleus of the egg fuses with that of the sperm, forming a diploid fertilised egg called the zygote.
- Oestrogen and progesterone levels are optimised during the ovulation cycle to create ideal circumstances in the uterus for implantation.
- When the embryo implants into the uterine wall, pregnancy occurs. Pregnancy is controlled by the secretion of progesterone by the corpus luteum in the first three months and later by the placenta.
- The hormone oxytocin promotes coordinated contraction of the smooth muscle of the uterus during labour, resulting in the birth of the baby.

CHECK YOUR UNDERSTANDING 2.3

1. Describe the formation of an egg cell (ovum) and its maturation.
2. What is a Graaffian follicle and what does it become once it has ruptured?
3. If fertilisation does not occur, what happens to the:
   - **a** Graaffian follicle
   - **b** ovum
   - **c** endometrium?
4. What is menstruation and when does this take place in the menstrual cycle?
5. Draw a labelled diagram to show the structure of a mature sperm cell.
6. Create a flow chart to represent the steps that occur during fertilisation.

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7 The flow chart and graph shown in figures 2.41 and 2.42 summarise hormonal control of fertilisation, implantation and pregnancy in humans. The graph (Fig. 2.42) represents hormone levels in females at various points in the menstrual cycle. Analyse information from the flow chart and the graph to complete the accompanying worksheet on hormonal control of reproduction.

Worksheet
Hormonal control of reproduction

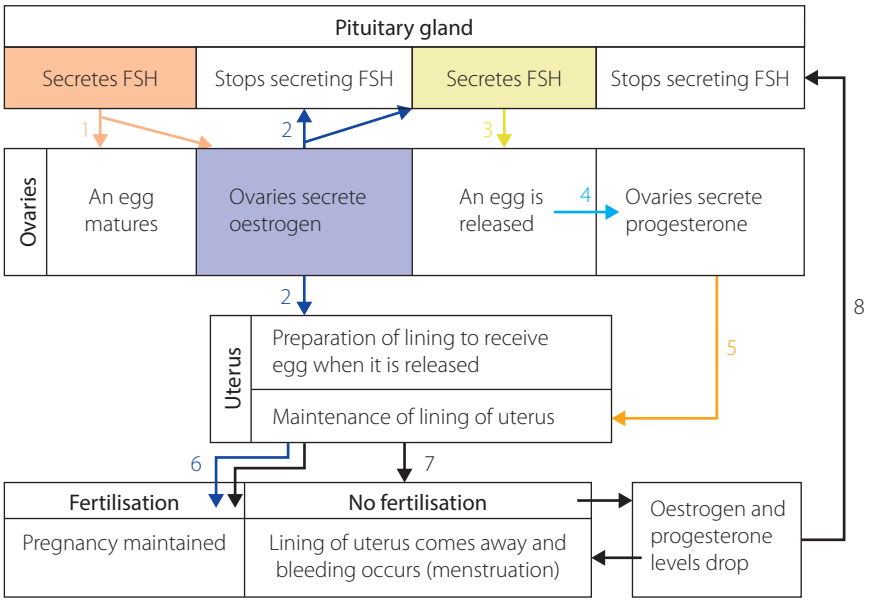

FIGURE 2.41 Hormonal control of reproduction in females

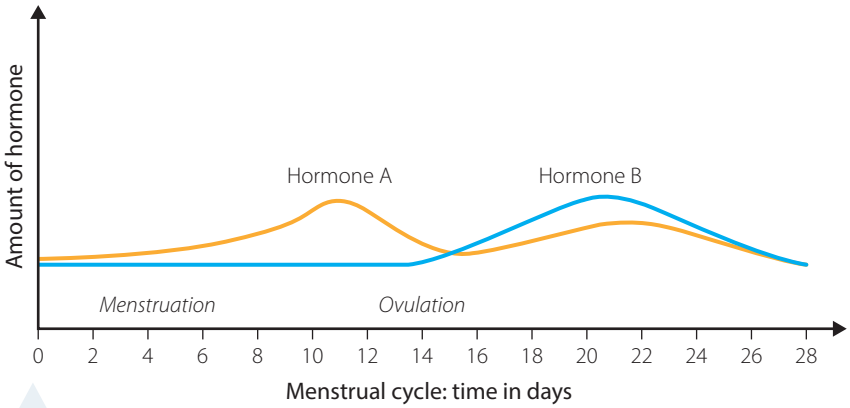

FIGURE 2.42 Hormone levels at different stages of the menstrual cycle

8 Using Figure 2.39 and the text on pages 68–9 as a starting point, create a flow chart depicting the control of pregnancy and lactation. You may need to add more arrows to the diagram and you will need to provide labels and annotations. Number the annotations to show the cyclical flow of hormone secretion in the order in which it occurs.

2.4 Manipulation of reproduction in agriculture

Agriculture is the cultivation and breeding of animals, plants and fungi for food, biofuels, fibre, timber and other products used to sustain and enhance human life. It includes both crop and livestock farming, forestry and fisheries and their products, to meet a wide range of human needs such as clothing, medicines, tools, furniture, artistic display and economic gain or profit.

Humans have been manipulating plant and animal reproduction for many hundreds of years. The techniques of manipulation have become more sophisticated as scientific knowledge of reproductive processes has advanced. The purpose of this human intervention is to improve the quality and yield of food. The term **reproductive technology** applies to any use of technology to assist and improve reproduction.

INVESTIGATION 2.3

IVF is dealt with in Chapter 9, together with other reproductive technologies.

A secondary source investigation of scientific knowledge and its application to reproductive technologies in agriculture

Worksheet
Human embryonic stem cell debate

BACKGROUND

Reproductive technologies range from those that manipulate fertilisation (such as artificial insemination, IVF, artificial pollination) to those that split up embryonic stem cells and involve embryo implantation (including embryo splitting and cloning, as well as embryo sexing and selection). Reproductive technology depends on scientific knowledge and an understanding of the reproduction process, such as control of seasonal breeding, hormonal regulation of ovulation and the oestrous cycle (Fig. 2.43), pregnancy and embryonic development.

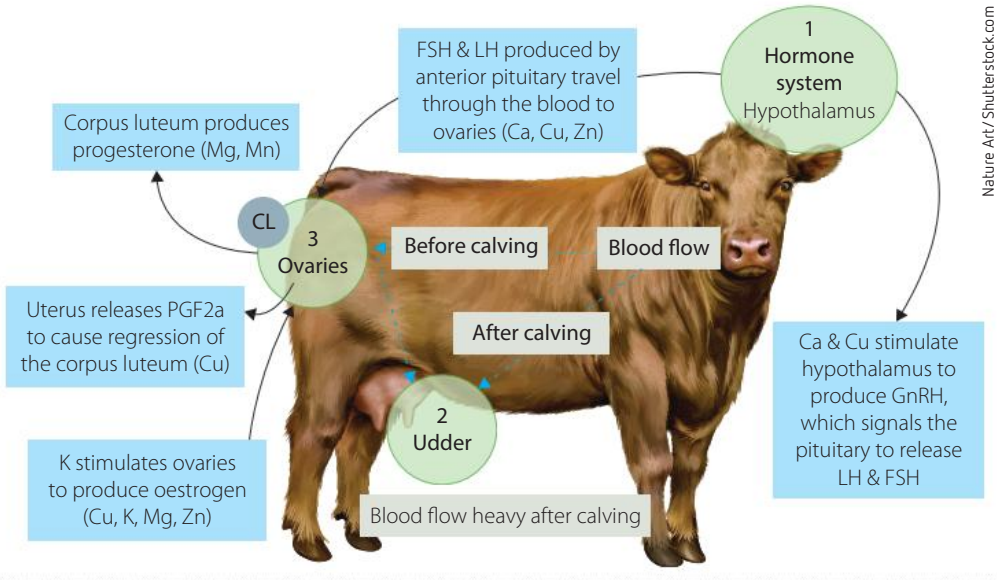

FIGURE 2.43
The role of hormones and minerals in controlling the oestrous cycle of a cow

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» The use of such technologies raises concerns about ethical issues such as stress on animals as well as environmental stress. The aim is to reduce these concerns while optimising the benefit for both farmers and consumers.

Ethical issues related to reproductive technologies are dealt with in more detail in Chapter 8.

Reproductive technologies and their use in agriculture are dealt with in more detail in Chapter 9.

AIMS

1. To investigate the reproductive techniques that have been implemented in agriculture as a result of advances in knowledge about reproduction in plants and animals
2. To analyse three techniques used to manipulate reproduction of plants and animals in agriculture
3. To evaluate the impact of this knowledge, taking into account any advantages, disadvantages and ethical concerns that result from these techniques

METHOD

Using examples such as the diagram in Figure 2.43, your own research and the information in this chapter, evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture, by finding out:

- *what* scientific knowledge was needed to make possible the specific manipulations of plant or animal reproduction that are now in use
- the *name* of the reproductive technique that was developed using this knowledge, and *when* the technique was introduced
- *how* reproduction is manipulated using this technique
- the *impact* of the scientific knowledge on the introduction of the technique. Include *advantages*, *disadvantages* and ethical issues.

Ethical understanding

Literacy

RESULTS

To present your results, copy and complete the table below.

SCIENTIFIC KNOWLEDGE ABOUT REPRODUCTION	REPRODUCTIVE TECHNIQUE (AND WHEN IT WAS INTRODUCED)	HOW REPRODUCTION IS MANIPULATED IN PLANTS AND/OR ANIMALS USING THIS TECHNIQUE	IMPACT OF KNOWLEDGE: ADVANTAGES AND/OR DISADVANTAGES (OR ETHICAL CONCERNS) OF THIS TECHNIQUE

EXTENSION

For students who wish to explore further, investigate the role of minerals in activating or promoting the action of hormones. Some examples are shown in Figure 2.43.

CONCLUSION

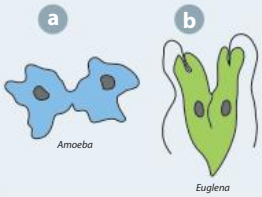
Write a few sentences summarising your findings about the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture.

2 CHAPTER SUMMARY

Sexual and asexual reproduction: How does reproduction ensure the continuity of the species?

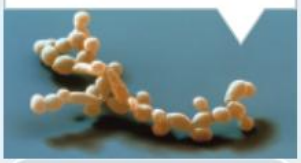
BINARY FISSION

In bacteria (*Listeria* sp.)

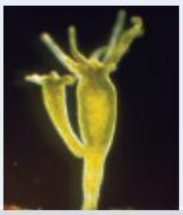


In protists (amoeba, euglena)

BUDDING



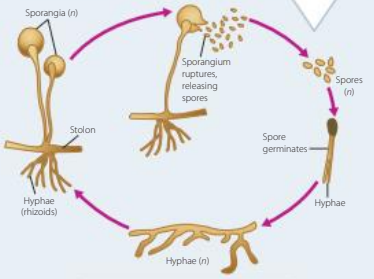
In fungi (yeast)



In animals (hydra)

Asexual reproduction: One parent, genetically identical offspring, well suited to survive in their enviroment

SPORES:



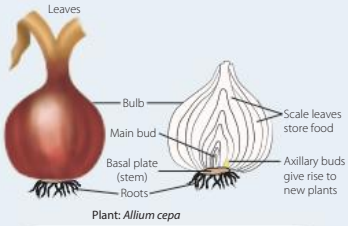
In fungi (mould)

PROPAGATION

Tuber

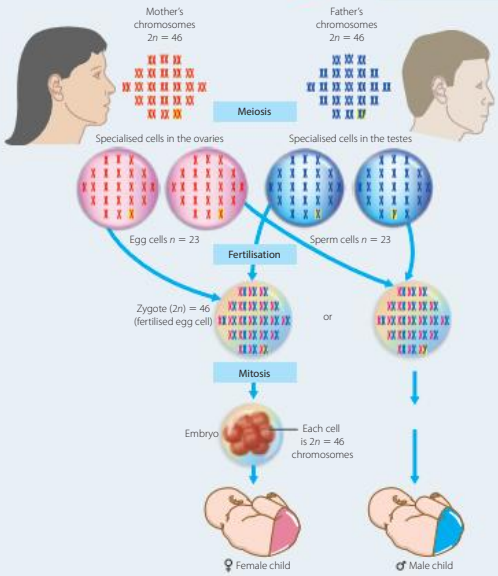
Rhizome

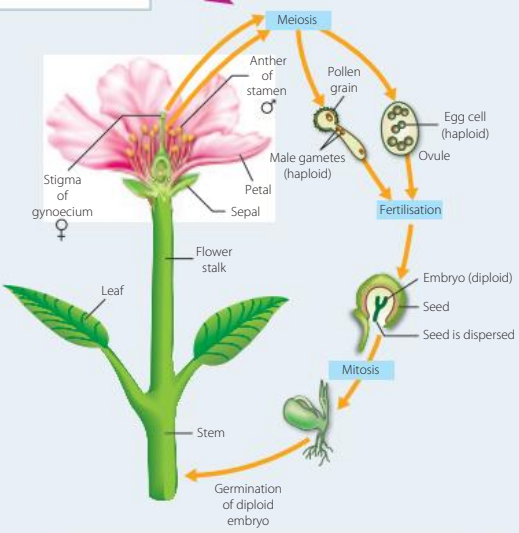
Apomixes



In plants (bulbs)

Sexual reproduction:
Two parents, offspring with genetic variability and new gene combinations that increase chance of survival in the face of sudden enviromental change





Meiosis and **fertilisation** in flowering plants

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Strategies for reproductive success: mechanisms to bring male and female cells together

Pollination in flowering plants

Pollination in flowering plants

External and internal fertilisations in animals

Large number of gametes mature and are released together

External fertilisation in coral

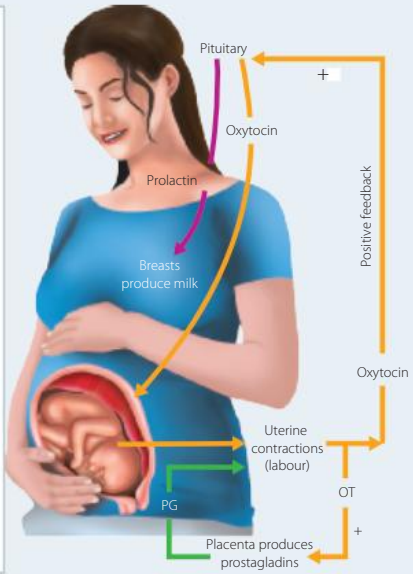
Smaller number of gametes assured of meeting; energy expended in finding a mate

Internal fertilisation in reptiles

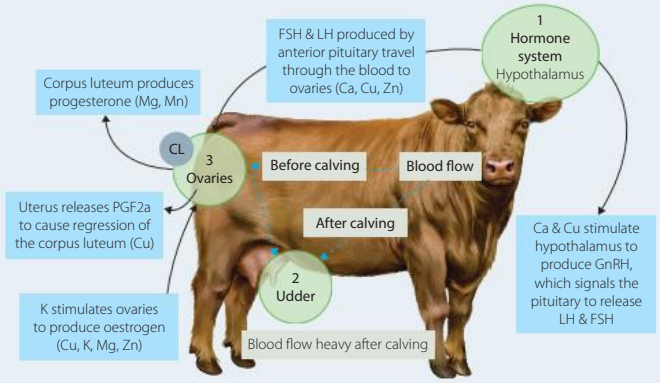
Hormonal control of the female reproductive cycle and maintenance of pregnancy and birth

Oestrogen and **progesterone** levels regulate *ovarian* and *menstrual cycles* and create ideal conditions for *ovulation*, *implantation* and maintaining *pregnancy*.

Oxytocin is released to stimulate uterine contractions and *birth*.



Manipulation of reproduction in agriculture



Scientific knowledge including understanding of seasonal breeding, hormonal regulation of ovulation and the oestrus cycle, pregnancy and embryonic development is necessary for manipulation of reproduction in agriculture.

2 CHAPTER REVIEW QUESTIONS

Review quiz

1. Distinguish between the following terms:
   - **a** sexual and asexual reproduction
   - **b** gamete and zygote
   - **c** eutherians, marsupials and monotremes
   - **d** fertilisation and pollination
   - **e** seed dispersal and seed germination
   - **f** budding and binary fission.
2. Using the examples of coral and amphibians, explain mechanisms that ensure male and female gametes can find each other when they are released, so that fertilisation can take place.
3. Explain why internal and external fertilisation are each suited to a particular type of environment.
4. Using a table, compare the advantages and disadvantages of internal and external fertilisation.
5. Explain how the relationship between the number of gametes produced and the type of fertilisation ensures continuity of the species.
6. Using a Venn diagram, compare the processes of sexual and asexual reproduction.
7. What is a hermaphrodite? Using an example, give one advantage and one disadvantage of hermaphroditism in ensuring the continuity of species.
8. Construct a table to compare oviparous, viviparous and ovo-viviparous development, using examples.
9. Describe two reproductive mechanisms in Australian animals to ensure the survival of the embryo.
10. Use examples to illustrate vegetative propagation of roots, stems and leaves as forms of asexual reproduction in plants.
11. Draw a sequence of diagrams to show:
    - **a** binary fission in bacteria
    - **b** budding in fungi
    - **c** binary fission and budding in protists
    - **d** spore production in bread mould.
12. Draw a scientific diagram of a longitudinal section through a flower and label the male and female reproductive parts.
    - **a** Write the function of each part next to the label.
    - **b** Draw arrows on the diagram to show how self-pollination occurs.
    - **c** Explain how self-pollination may reduce the chances of the continuity of species under some conditions, but increase the chances under other conditions.
13. Using words and arrows, construct a flow chart to show each step in the process of sexual reproduction in a flowering plant.
14. In a table, compare reproduction by means of spores in ferns and seeds in flowering plants.
15. Use a Venn diagram to compare wind, bird and insect pollination in flowering plants.
16. Analyse asexual reproductive mechanisms in plants, fungi and protists, and explain how these mechanisms ensure continuity of the species.
17. Distinguish between the terms:
    - **a** seasonal breeders and continuous breeders
    - **b** gonads and gametes
    - **c** ovulation and menstruation
    - **d** progestogen and progesterone.
18. Explain why offspring produced by asexual reproduction are similar to their parents, whereas offspring produced by sexual reproduction differ from their parents.
19. Why does DNA need to replicate before cell division?
20. Describe the journey of a human ovum from the ovary to when it is fertilised.
21. Use words and arrows to describe the journey of a sperm until it reaches the ovum.
22. Draw a diagram of a sperm and an egg at the exact point in time that fertilisation occurs. Nuclear detail is important.
23. Describe the hormonal control of gamete production and fertilisation.
24. Explain why, during the reproductive cycle, ovulation cannot occur in a pregnant female.
25. Name two hormones secreted by:
    - **a** the pituitary gland, that act on the ovary in human reproduction, and describe their action
    - **b** the ovary, that act on the uterus of a female during sexual reproduction.
26. Create a flow chart to model the hormonal control of pregnancy in human reproduction.
27. Referring to the graphs in Figure 2.35, explain the relationship between FSH, LH, oestrogen and progesterone in:
    - **a** bringing about ovulation
    - **b** bringing about menstruation.

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28 Explain the mechanisms in human reproduction that ensure:

- **a** sperm find the egg cell
- **b** only one sperm fuses with the egg.

29 Construct a table to summarise the hormones involved in the control of pregnancy and birth, and the functions of these hormones.

30 Create a brochure for a healthcare facility to explain *two* of the following:

- **a** the role of the placenta in supporting foetal development. Explain the necessity for correct hormone levels
- **b** the biological process of birth, including an explanation of hormonal regulation
- **c** the time of the month when a female is most fertile and ovulating, including an explanation of hormonal levels
- **d** the conditions necessary for implantation of an embryo and maintenance of pregnancy in a female, including the influence of hormone levels
- **e** the conditions necessary for healthy sperm production in a male, including an explanation of hormonal levels.

31 Using examples, explain why farmers rely on vegetative propagation as a form of asexual reproduction in plants.

Exam preparation

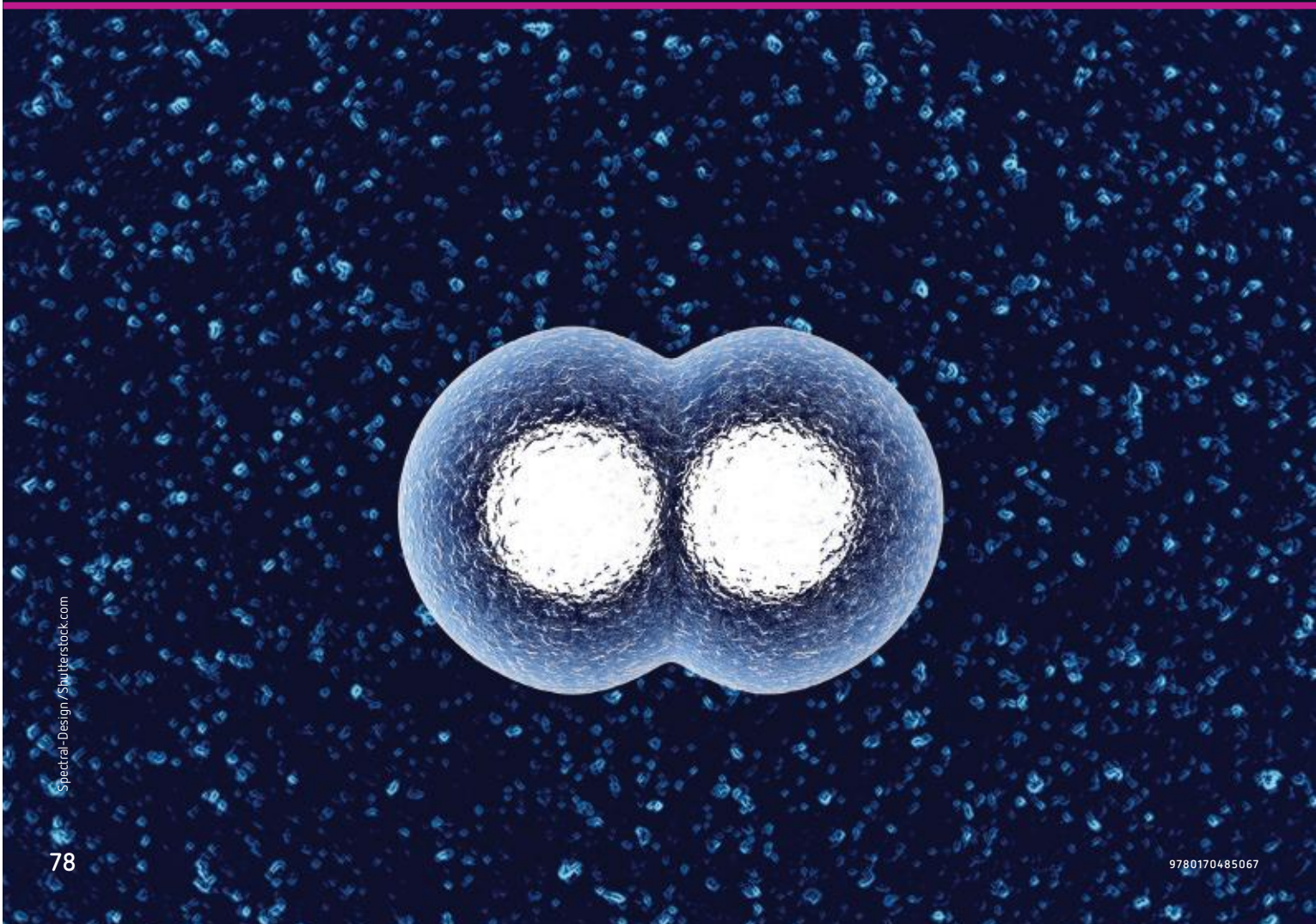
3 Cell replication

INQUIRY QUESTION

How important is it for genetic material to be replicated exactly?

Students:

- model the processes involved in cell replication, including but not limited to:
  - mitosis and meiosis (ACSBL075) CCT ICT
  - DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding (ACSBL076, ACSBL077)
- assess the effect of the cell replication processes on the continuity of species (ACSBL084) ICT



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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
3.1 A practical and secondary-source investigation of mitosis
3.2 Modelling DNA replication using the Watson and Crick model
3.3 Literature review of the effect of cell replication processes on the continuity of species

Worksheets
- The discovery of DNA
- Chromosomes - keys to inheritance
- The structure of nucleic acids
- Evaluating resources

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

Have you ever wondered why it is so important for genetic material to be replicated exactly? Each living creature has a different genetic code that carries instructions for its own production. How do the genetic instructions for wombats differ from those for koalas, dogs, penguins or even mushrooms? Parents give rise to offspring of the same species, with a similar genetic makeup to their own, so they can survive in the same environment. It is essential that organisms pass on their own unique genetic instructions or information from one generation to the next, to ensure the continuity of the species.

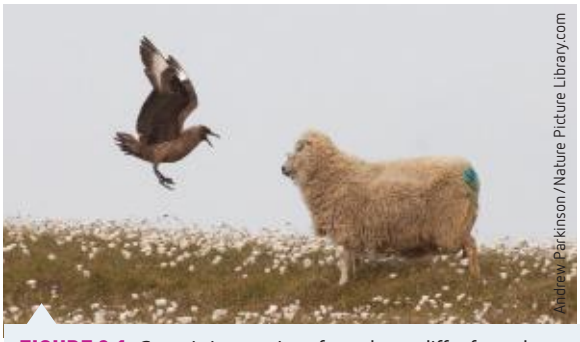
Andrew Parkinson / Nature Picture Library.com

FIGURE 3.1 Genetic instructions for a sheep differ from those of a bird, grass or flowers.

How can something as tiny as a cell carry all the instructions needed to make a large, complex, living organism? When scientists began searching for the 'code of life' in cells, they realised that the genetic code would need to be in molecular form to be small enough to fit all the instructions into a cell. As chromosomes are composed of DNA (deoxyribonucleic acid) and protein, scientists deduced that the genetic code must be carried by one of these two macromolecules – either in proteins or in DNA.

It was expected that the genetic code would be complex, because it needs to store large amounts of information accurately over long periods of time. The genetic code would also need to be copied exactly and any errors that arose during copying would need to be easily fixed. Most scientists favoured protein as the carrier of the genetic instructions, because proteins are complex and varied, being made up of at least twenty different amino acids.

In 1953, James Watson and Francis Crick discovered the structure of DNA and how it held the 'code of life'. What surprised most biologists was that genetic information could be carried by a molecule that uses an 'alphabet' made up of only four 'letters': the bases adenine (A), cytosine (C), guanine (G) and thymine (T). In the decade following 1953, the combined work of many scientists led to an understanding of how the information in DNA can lead to specific proteins being produced.

In this chapter, you will find out how Watson and Crick worked out the structure of DNA, as well as when and how genetic material is replicated and why it is so important for replication to be exact.

3.1 Cell division – mitosis and meiosis

Why do cells need to divide? What mechanisms are in place for the faultless transmission of genetic information from parent to daughter cells during division? When a cell divides, how does every daughter cell get an exact copy of the genetic instructions? As you study the mechanisms of cell division, you will discover how this is achieved.

There are two types of cell division: mitosis and meiosis.

In unicellular protist organisms, cell division by asexual binary fission is common – one organism becomes two and no sex cells or gametes are involved. In multicellular organisms, cell division by **mitosis** leads to the formation of two new identical cells that contribute to the growth of the organism.

Meiosis is a different type of cell division. It gives rise to gametes that transmit genetic material from one generation to the next during sexual reproduction. Meiosis will be dealt with in more detail in Chapter 5.

The role and importance of mitosis

Mitosis plays an important role in:

- growth of multicellular organisms – from fertilised egg to embryo, from infant to adult, growth relies on mitotic division followed by cell assimilation, enlargement and differentiation
- repair of damaged tissue and replacement of worn-out cells
- asexual reproduction – for example, growth of plants from cuttings, and the cloning of organisms
- genetic stability – mitosis ensures the precise and equal distribution of chromosomes to each daughter nucleus, so that all resulting cells have the same chromosome number and genetic information as each other and the original parent cell.

See Chapter 4, page 129 for information on stem cells and gene expression.

Most multicellular organisms start life as a single fertilised egg cell or **zygote**, which grows into an embryo. Embryonic cells are able to divide repeatedly and are **pluripotent** (that is, they have the potential to develop into any type of tissue) (Fig. 3.2). These cells are termed **embryonic stem cells**. You may have heard of these already, because the use of embryonic **stem cells** is a subject that often draws media attention and discussion among people. This is because the use of these cells in therapeutic medicine and for research involves the destruction of the embryo, a contentious issue. This will be dealt with in greater detail in Chapter 8.

In mature organisms, not all cells continue to divide. As cells differentiate and specialise, they form tissues, and some of these cells lose the ability to divide by mitosis. Dividing cells remain at particular locations within the adult body; for example, dividing cells in the basal layer of the skin replace dead surface skin cells, and cells in bone marrow give rise to blood cells. **Adult stem cells** occur in almost every type of tissue but they are pre-specialised; for example, brain stem cells can only make brain tissue and heart stem cells can only make cardiac tissue. Cells in bone marrow can give rise to all types of blood cells, and so they are termed 'multipotent'. These adult stem cells can therefore not be used as widely in all types of tissue regeneration as pluripotent embryonic cells. Researchers are continuing to investigate how adult stem cells can be made pluripotent, so they can be used in tissue and organ regeneration. Unfortunately, reversing pre-specialisation of cells in tissue culture tends to make cells turn cancerous, so there is still much research to be done in this area.

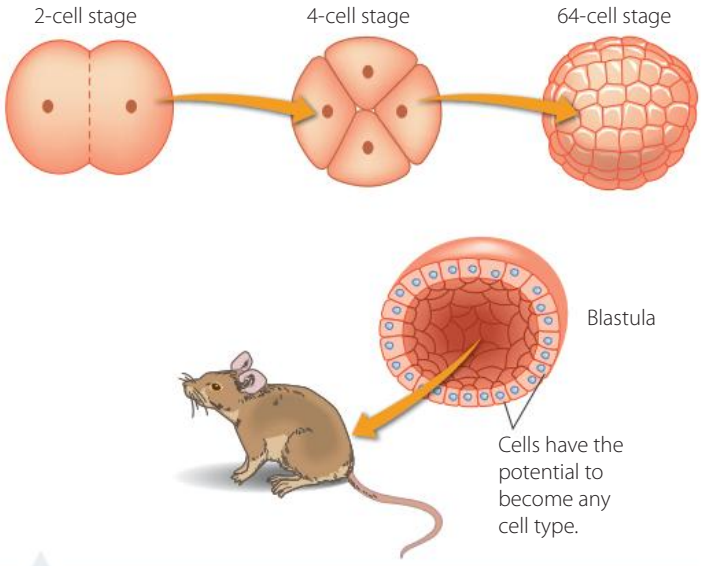

FIGURE 3.2 Early stages of mammalian embryonic development: mitotic division of pluripotent embryonic stem cells

In plants, tissue that can divide to form any other tissue is known as **meristem** and occurs at particular locations in the plant, such as the tip of the stem and the tip of the root.

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The role and importance of meiosis

Refer to Chapter 2 for information on asexual and sexual reproduction, and to Chapter 9 for information on cloning.

Meiosis is the type of cell division that occurs in the sexual reproductive organs of a plant or animal, and results in the formation of gametes (sex cells).

During sexual reproduction, two parents are involved in passing genetic material to the offspring. To prevent the chromosome number from doubling in each successive generation, a mechanism must exist to ensure that each parent contributes only half of his or her chromosomes to the new offspring. This mechanism is meiosis, a type of reduction division in cells in the reproductive organs of both plants and animals. Meiosis ensures that the chromosome number of each species is maintained (not doubled) during sexual reproduction (Fig. 3.3).

When a cell divides by meiosis, DNA replicates before division. The cell then undergoes two successive divisions:

- meiosis I, where the diploid cell divides into two haploid cells and the chromosome number is halved
- meiosis II, where the two cells each divide again, resulting in four haploid daughter cells (called a **tetrad**).

Each daughter cell has half the original number of chromosomes that the parent cell had.

These resulting daughter cells, or gametes in plants and animals, are:

- *egg cells* (in ovaries) and *sperm cells* (in testes) in animals
- *pollen grains* (in anthers) and *egg cells* (in ovules) in seed-producing plants.

Gametes are often referred to as 'vehicles of inheritance' because they carry genes from one generation to the next.

Meiosis is also the process by which genetic variation is introduced into a species. The process of meiosis and its contribution to genetic variation is dealt with in detail in Chapter 5.

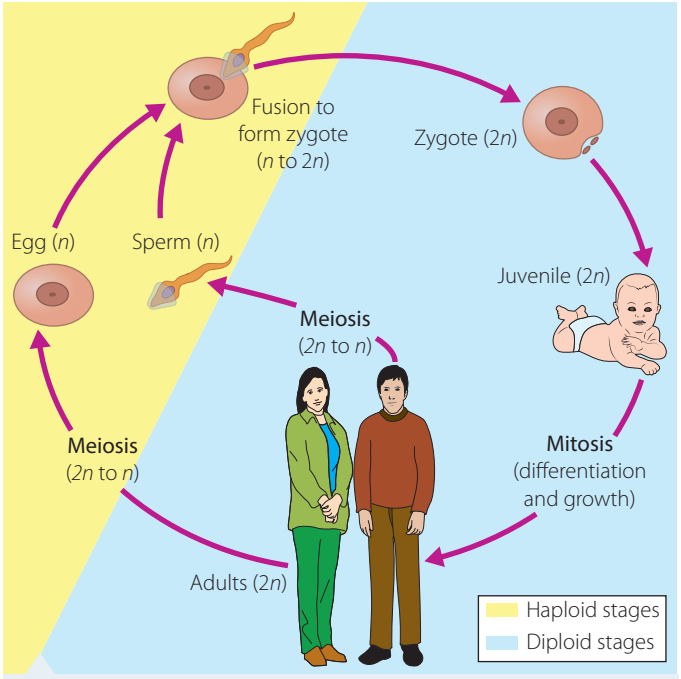

FIGURE 3.3 Life cycle of humans showing the importance of meiosis (formation of haploid gametes) and mitosis (differentiation and growth)

Cell division: why does DNA need to replicate exactly?

An organism's genetic code contains instructions for every feature of the individual, from its biochemistry, which allows it to function properly, to its physical features and body size. The genetic code even holds instructions for how its own genes are expressed and therefore for how all biological information in the organism is read, processed and converted (translated) from its coded form (DNA) to its end product (protein).

When a cell divides into two, it is vital that the DNA passed on to each daughter cell is an exact copy, so that each generation of cells contains the same genetic instructions. In unicellular organisms, the genetic code passed from parent cells to offspring maintains the genetic stability of the species. In multicellular organisms, all cells making up the body must contain the same genetic code so that they can function in a controlled and coordinated way.

Along the chromosome are long sequences of base pairs in specific regions of DNA that form units called genes. Genes code for the production of proteins, which in turn make up a large proportion of the structure of cells and also regulate the functioning of cells. For example, many body parts such as bones, hair, skin, muscles and connective tissue contain proteins, which are coded for by genes. Some proteins are enzymes (as you learned in Year 11), and these carry out and control all cell functions. Each step of every biochemical reaction in cells, such as chemical respiration and photosynthesis, is controlled by enzymes.

It is therefore vital for the functioning of cells and the stability of species that the genetic code is passed on without error. During mitosis, DNA is replicated precisely and the copies of all genes are divided up equally between the resulting cells. The cells are therefore clones of each other. In exploring the process of mitosis, you will see how this happens in somatic (body) cells.

All cells need to make particular end-products and function in a coordinated manner. DNA codes for proteins used inside the cell, as well as proteins such as hormones and enzymes that are exported and coordinate functioning or direct protein synthesis in other cells. A change in DNA, known as a **mutation**, may change the end product and, as a result, may have a harmful effect on the cell, by interfering with its structure and/or functioning. Sometimes mutations have no effect, and occasionally they may be beneficial. You will learn more about mutations and genetic variation in chapters 5 and 7.

KEY CONCEPTS

- *Binary fission* is cell division that occurs in prokaryotes for asexual reproduction.
- *Mitosis* is the nuclear division of eukaryotic cells for asexual reproduction (in unicellular organisms and propagation in plants) and for growth and repair (in multicellular organisms).
- *Meiosis* is the nuclear division of eukaryotic cells for gamete formation in sexual reproduction.
- Many protists, plants and fungi can reproduce by mitosis (asexual reproduction).
- Mitosis in multicellular organisms is a mechanism for tissue growth, maintenance and repair.
- Meiosis is the type of cell division that occurs in the sexual reproductive organs of a plant or animal and it results in the formation of gametes (sex cells) with the haploid number of chromosomes. It also introduces genetic variation into a population.
- In 1953, James Watson and Francis Crick discovered and modelled the double-helical structure of DNA.
- The sequence/order of the bases A, T, C and G in DNA holds information in a coded form.
- DNA replication is vital for the continuation of a species, as it allows an organism to reproduce its genetic code and pass it on.

The cell cycle

Weblink
Cell cycle
Play the animation and do the quiz to check your understanding.

Weblink
Cell cycle game

Weblink
Cell cycle animations
Watch the five short animations on the cell cycle and cancer.

Cell division and enlargement occur in a repetitive sequence called the **cell cycle** (Fig. 3.4). Mitosis is only one part of this cycle and usually takes about an hour or two. A complete cell cycle in actively dividing cells may take about 18–22 hours. The cell spends a large amount of time preparing for division (a stage called **interphase**, which precedes mitosis).

The cell cycle has four main phases.

- G_1 is a gap phase for *cell growth* before DNA replication. During this phase, cell enlargement takes place. Metabolic changes prepare the cell for division and the cell reaches a point at which it is committed to division, then enters the next phase.
- S is a *synthesis phase* during which DNA replicates – that is, the DNA in the cell is copied so that each dividing cell has two identical copies at the start of mitosis. At the end of cell division, one full copy ends up in each resulting daughter cell.
- G_2 is a second gap phase after replication, when enzymes in the cell *check the duplicated chromosomes* for any errors and correct these, and the cytoplasmic materials accumulate in preparation for division.
- Mitosis (*division of the nucleus*) then occurs, followed by **cytokinesis** (*division of the cytoplasm*). Cytokinesis marks the separation of one cell into two.

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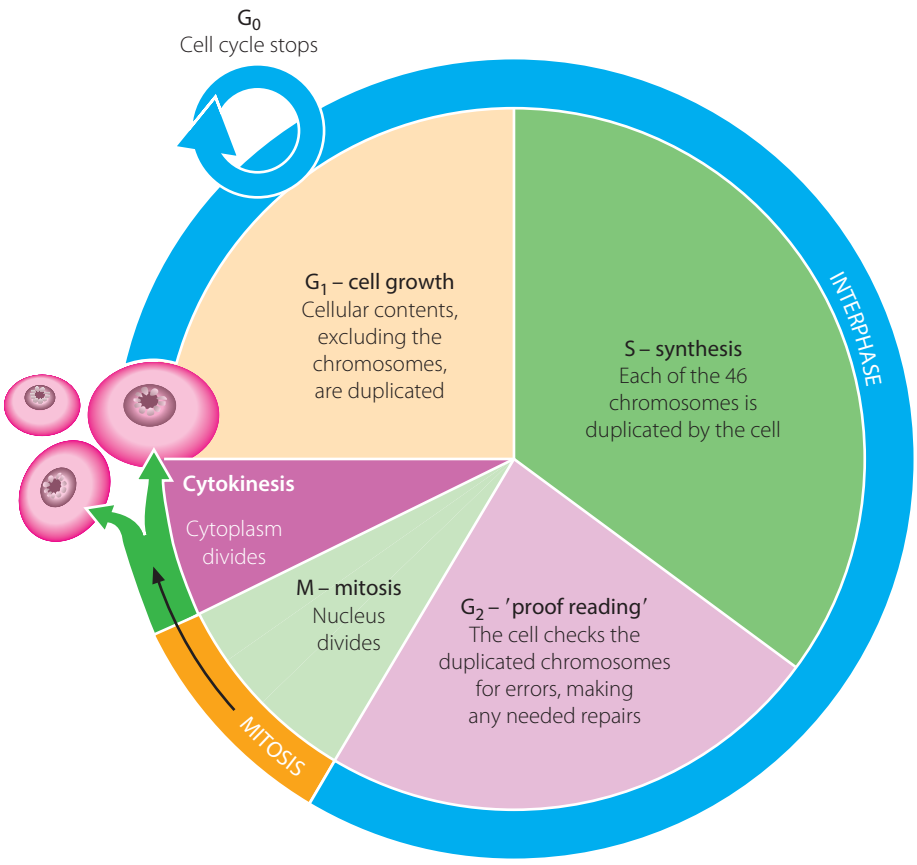


FIGURE 3.4 The cell cycle

In actively dividing tissue, following cytokinesis, each cell will enter G_1 again to repeat the cell cycle. This begins with a period of assimilation and cell enlargement, where cells increase in size by assimilating new materials into the cell boundaries and cytoplasm. This new material is made from the nutrients that an organism acquires. For example, these nutrients are the end products of digestion in animal cells and of photosynthesis in plant cells.

Not all cells continue to go through cyclical divisions. The basal layers of cells that need to be replaced often, such as skin cells and cells lining the digestive tract, divide rapidly. Some adult cells do not divide frequently and others, such as nerve cells, may last a lifetime. These cells are said to be terminally differentiated. They are in a phase called the G_0 phase and only re-enter the cell cycle under special circumstances.

Weblink
Cell cycle revision
Explore in more detail the cell cycle and mitosis, explained in diagrams and text.

Mitosis: division of the nucleus

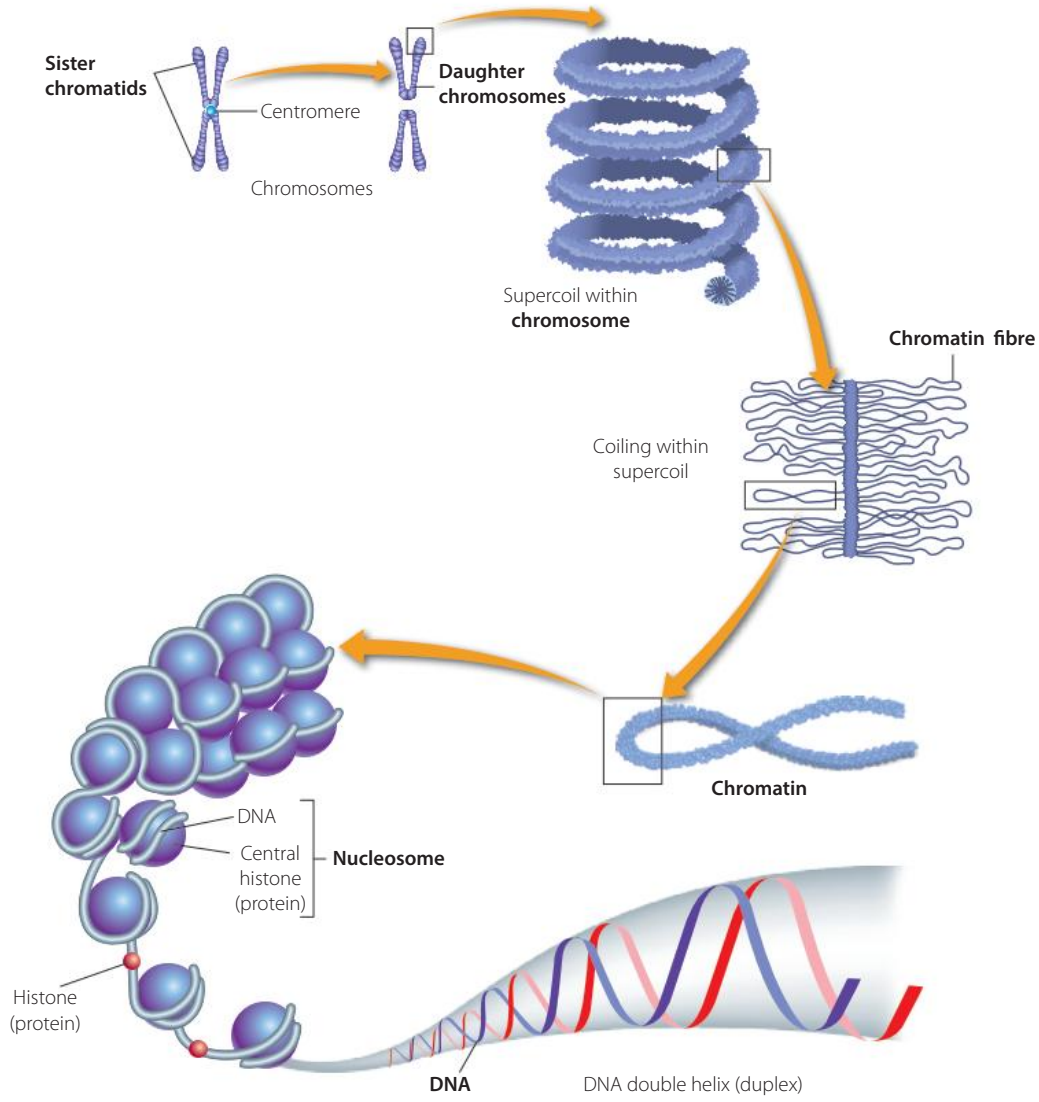
Mitosis is the type of nuclear division that ensures that daughter cells receive the same number and form (exact copies) of chromosomes as the parent cell. Therefore, before a parent cell can divide it must make an accurate and complete replica of the genetic information encoded in its DNA. This needs to be accurately and equally distributed to the resulting daughter cells. Mitosis is a gradual and continuous process, but is usually described in four phases: *prophase*, *metaphase*, *anaphase* and *telophase*.

In order to understand the stages of mitosis, you need to be familiar with the following concepts:

- In a non-dividing cell, the DNA wound around protein is known as **chromatin** (Fig. 3.5).
- Chromosomes contain linear sequences of *genes*, the units of **heredity** that code for the inherited characteristics of an organism; for example, there are genes on human chromosomes that determine eye colour, hair colour and height.

- Each organism has a set number of chromosomes (for example, humans have 46 chromosomes, a platypus has 52 and a lettuce has 18).
- During the S phase, chromosomes replicate and the two chromosome copies or **chromatids** are held together by a structure called a **centromere**. These **sister chromatids** (Fig. 3.5) separate and move to opposite poles during mitosis. They are now known as **daughter chromosomes** and are distributed into two daughter nuclei. Following this, the cytoplasm divides, separating the two daughter nuclei from each other (cytokinesis).

FIGURE 3.5
Structure of a chromosome at the start of mitosis



Stages of mitosis

At the start of mitosis, DNA is so condensed or highly folded that the chromosomes are visible with a light microscope.

Although mitosis is a continuous process, it is easier to understand if we divide it into identifiable stages. There are four main stages of nuclear division in mitosis: following *interphase* (the S phase during which DNA replication takes place) are *prophase*, *metaphase*, *anaphase* and *telophase*. The late stages of nuclear division are accompanied by the start of cytokinesis. The process of cell division is summarised in Table 3.1. To keep the diagrams simple, only two pairs of chromosomes (four chromosomes in total) are shown in each cell.

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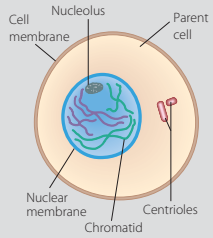
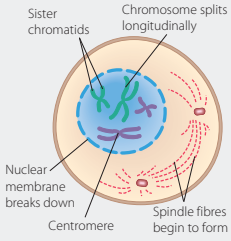
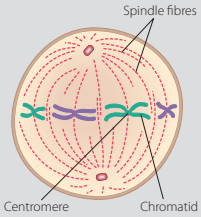
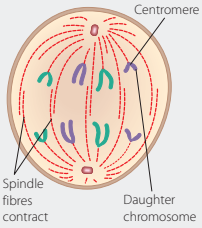
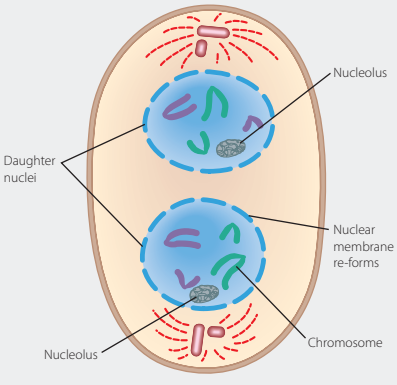
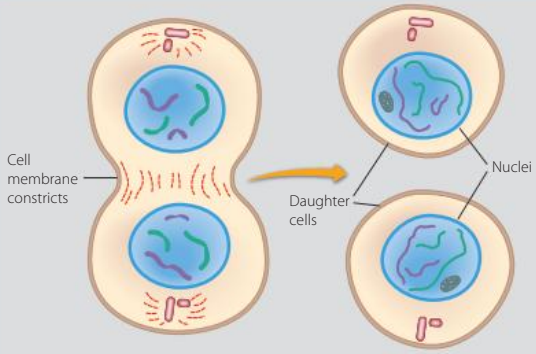
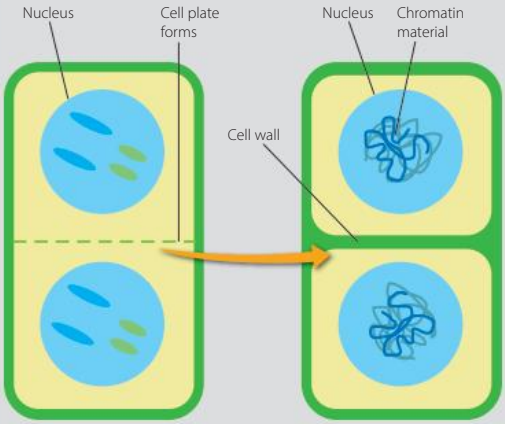
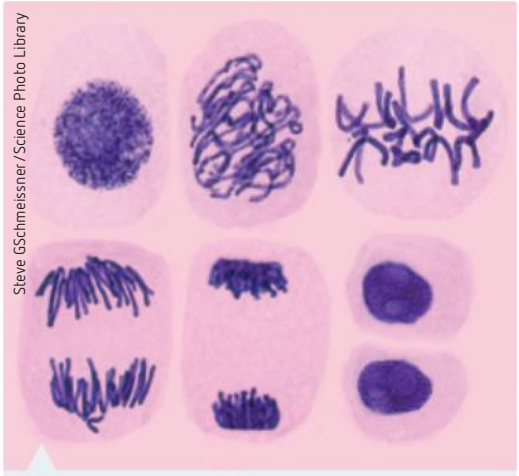
TABLE 3.1 Mitosis in an animal cell

STAGE		DESCRIPTION	DIAGRAM
Interphase / early prophase		Interphase occurs in the S phase of the cell cycle, where DNA synthesis occurs: DNA replicates (makes an identical copy of itself). It appears diffuse and spread out and is termed 'chromatin'. It is not yet recognisable as individual chromosomes. In early prophase (seen in this image), DNA begins to separate into chromosomes.	 G_2, early prophase: DNA replication complete
Mitosis	Prophase	In prophase, the chromatin material shortens and thickens by coiling and the DNA separates out into chromosomes, which are now visible with a light microscope. Each chromosome contains two copies of the DNA. Each copy is called a sister chromatid and these are joined by a single centromere. The nuclear membrane begins to break down and is no longer visible; a spindle begins to form from the broken-down material and extends across the cells (like the imaginary lines of longitude that are drawn on a globe of the world).	 Prophase: chromosomes condense, become visible and spindle apparatus forms
	Metaphase	In metaphase, the chromosomes line up across the centre or 'equator' of the cell, each attached to the spindle fibres by a centromere. Each chromosome consists of two identical sister chromatids.	 Metaphase: chromosomes align along the equator of the cell
	Anaphase	Anaphase begins when proteins in the centromere are cleaved, which allows the sister chromatids to separate. Each chromatid becomes a chromosome. The spindle fibres contract and the chromosomes are pulled by their centromeres to opposite ends of the cell. The spindle fibres contract and, as the sister chromatids begin to separate, they are now known as daughter chromosomes. They are pulled towards opposite poles of the cell and their movement is assisted by the centromere.	 Anaphase: sister chromatids separate to opposite poles of cell
	Telophase	The daughter chromosomes gather at opposite poles of the cell. The spindle breaks down. The nuclear membrane and nucleolus reappear. Nuclear division or mitosis is now complete. The result is two nuclei with chromosomes identical to each other and to the original nucleus in the parent cell. The nuclear membrane forms around the two nuclei, now called daughter nuclei. Mitosis is now complete.	 Telophase: nuclear membranes assemble around two nuclei

STAGE	DESCRIPTION	DIAGRAM
Cytokinesis	Division of the cytoplasm occurs, separating the two daughter nuclei so that each is in its own cell. Cytokinesis differs in plant and animal cells. • Animal cells: the cytoplasm constricts in the centre of the cell between the two daughter nuclei, and 'pinches off' (similar to squeezing a round balloon in the centre until the edges meet and then giving it a twist to separate it into two bubbles).	 Cytokinesis in animal cells: division of cytoplasm into two; G_1, cell growth of daughter cells
	• Plant cells: a cell plate forms while the nucleus is still in telophase; thickenings appear on the spindle fibres in the region of the equator and join up to form a cell plate made of pectin compounds. Cellulose is then deposited on either side, forming a cell wall to separate the two daughter nuclei.	 Cytokinesis in plant cells



Steve GSchmeissner/Science Photo Library

FIGURE 3.6 Root tip cells undergoing mitosis and cytokinesis, showing mitotic stages (in order from top left to bottom right) interphase, prophase, metaphase, anaphase, telophase and cytokinesis. Chromosomes have been stained with toluidine blue.

Weblink
Mitosis poster and video
Download or print the poster of mitosis, then scroll down and watch the video clip of live plant cell mitosis using time lapse photography

Weblink
Narrated video – mitosis
Watch the video clip of mitosis, presented diagrammatically

Weblink
Interactive animation – mitosis

Cytokinesis

Cytokinesis is the final step in cell division (Table 3.1, Fig. 3.6). It is the division of the cytoplasm and begins while the nucleus is completing its division. Cytokinesis is important because it separates the newly formed daughter nuclei and ensures that each cell has only one nucleus. The outcome at the end of mitosis and cytokinesis is two daughter cells with chromosomes that are identical to each other and to the original parent cell. The daughter cells then enlarge until they are the same size as the original adult cell (during G_1, assimilation as well as cell enlargement occurs). The nucleus of each cell controls all cell activities. The ratio between the proportion of nucleus and cytoplasm remains constant. If the cytoplasm exceeds a certain proportion of the cell, the ability of the nucleus to control its functioning decreases and this may help trigger cell division.

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Telomeres and ageing

As people age, some of the cells in their dividing layers reach a point where they can no longer divide. Scientists have discovered that there is a change in the structure of the ends of the chromosomes in these cells. A telomere is a DNA–protein region at each end of a chromosome that seems to function in preventing chromosomes from sticking to each other (Fig. 3.7). Telomeres seem to have additional functions in extending the life of a cell, such as protecting the genome from degradation and unnecessary recombination and repair. Children have longer telomeres than older people. With successive divisions, a small amount of DNA is lost from the telomeres in a cell and they become shorter. Once the telomeres reach a certain predetermined length, the cell stops dividing and this leads to cell senescence and/or death. There seems to be variation in the length of telomeres that people are born with and in the rate at which they shorten. Lifestyle choices may also influence telomere shortening. This has become an interesting area of research into ageing and disease.

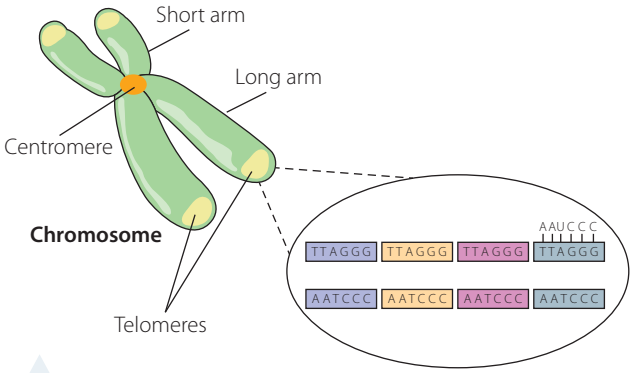

FIGURE 3.7 A telomere is a region at the end of a chromosome that shortens as we age.

INVESTIGATION 3.1

A practical and secondary-source investigation of mitosis

INTRODUCTION

Mitosis can be observed relatively easily in cells in the growing region of a root tip. In prepared slides, these cells have been dyed using a stain (such as acetic orcein or a counterstain with toluidine blue) that is taken up specifically by chromosomes.

Because the tissue in the prepared slide is no longer living, you will see the cells in the stages of division that they were in when the root tip was killed with a fixative.

The division of cells at any one time is not synchronised – different cells are in various stages of division. The phase that is most commonly represented in the slide is the phase that cells remain in for the longest period of time. Review the cell cycle and predict which phase you would expect this to be.

Using the formula for the **mitotic index**, you can calculate the proportion of cells in the field of view that are undergoing mitosis compared with the number not undergoing mitosis (and instead are in the interphase or growth phase).

The prepared slides that you will look at show stained tissues that are no longer living. Modern advanced microscopy techniques (not usually available in schools) would allow you to view and record living tissue that is actively dividing. Videos taken using this technology are available for viewing on the Internet, often on YouTube. You are encouraged to view video clips taken in real time of cells dividing. Record all the references that you use.

AIM

To gather information on the sequence of changes in the nucleus of plant cells undergoing mitosis

PREDICTION

Review the cell cycle and predict which phase you would expect to see the most number of cells in.

MATERIALS

- compound light microscope
- prepared slides of onion root tip

RISK ASSESSMENT

Complete the table to assess the risks and outline how you can safely manage these.

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?
Microscope		
Glass microscope slides		

METHOD

Revise microscope safety and risk assessment from *Biology in Focus Year 11*.

1. Using correct microscope technique, observe a prepared slide of a root tip using the compound light microscope.
2. Under low power, locate and identify the apical meristem. This is found just behind the root cap and is easily identified by numerous small cells that appear square in shape and have nuclei that are large relative to the whole cell area. Look for cells with darkly stained chromosomes, indicative of mitosis occurring. Move the slide so that these are in the centre of your field of view and then change to high power to observe them in detail.
3. Under high power, identify cells in at least four different stages of mitosis. Use figures 3.6 and 3.8 to assist you.

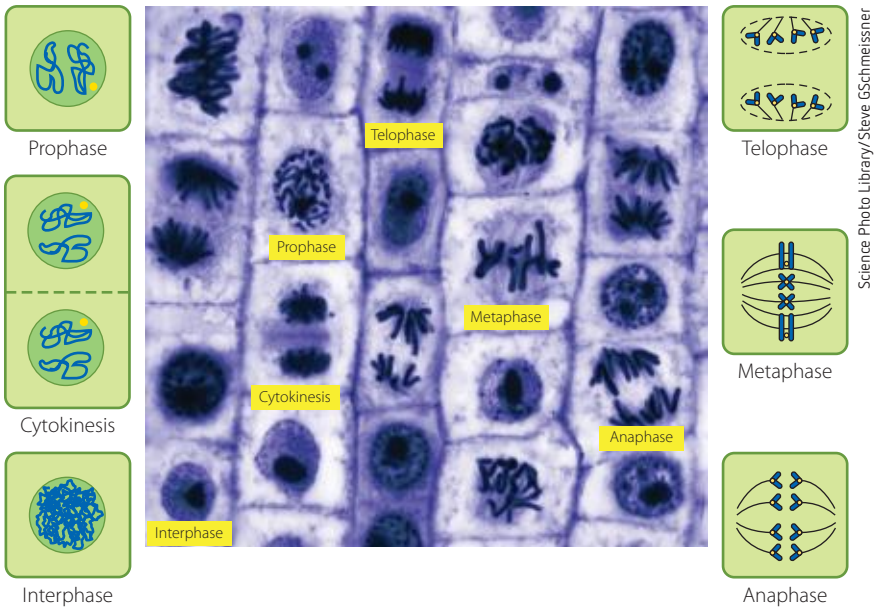

FIGURE 3.8
A photomicrograph of onion root tip cells undergoing mitosis, with examples of the different phases highlighted

RESULTS

1. Record your results in the form of fully labelled diagrams. Remember to include a heading for each diagram and state the magnification used. Write a brief description of what is happening at each stage in the diagram.
2. You may wish to take photographs of your field of view using your mobile phone. Print the photograph and label on it the various stages of cell division that you can see.
3. Calculate the mitotic index of your field of view (refer to Introduction) using the formula:

Numeracy

$$\text{Mitotic index} = \frac{\text{number of actively dividing cells in field of view}}{\text{total number of cells in field of view}}$$

4. Write a practical report using a procedure text type, under the headings Aim, Risk assessment, Materials, Method, Results and Conclusion.

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DISCUSSION

Discuss the accuracy, reliability and validity of data you gathered during the investigation. Estimate the uncertainty of measurements in your data. (See Chapter 1, page 20 for estimating uncertainty.)

CONCLUSION

Write a summary statement that relates to the aim and your prediction in this investigation, including a brief outline of what you found.

EXTENSION

Carry out one of the following optional extension activities.

1. Find a video clip of mitosis filmed under a light microscope or fluorescence microscope in real time. Record the weblink so that your peers can view it.
2. Estimate the duration of each stage of mitosis by recording data on the observed frequencies of each stage of cell division in your field of view. Present your data in table format, using Table 3.2 as a guide.

TABLE 3.2 Observed frequencies of stages of cell division in a root tip field of view

	OBSERVED OCCURRENCE IN REPRESENTATIVE MICROSCOPE FIELDS		DURATION	
STAGE OF CELL DIVISION	NUMBER OF CELLS	% OF TOTAL CELLS	% OF TOTAL TIME	ESTIMATED TIME (MINUTES)
Interphase				
Prophase				
Metaphase				
Anaphase				
Telophase				
Cytokinesis				

Weblink
Mitosis animation and quiz
Work through the quiz to check your understanding.

Replication of DNA outside the nucleus

Although each cell has one nucleus, it will have many organelles such as mitochondria and chloroplasts in the cytoplasm. The number of each type of organelle depends on the functions of the tissue. Cells that require large amounts of energy have many millions of mitochondria. Photosynthesising cells in the leaves of plants have many chloroplasts. Just as cells divide, these organelles must also divide to maintain their numbers for normal tissue functioning.

A small amount of DNA is located in organelles such as mitochondria and chloroplasts, in the cytoplasm. This DNA, called **non-nuclear DNA** or extrachromosomal DNA, carries genes that are important to cell metabolism.

In cytokinesis, when the cytoplasm divides in half, organelles in the cytoplasm such as mitochondria and chloroplasts reduce in number per cell, as they are now distributed between two daughter cells. To avoid a repeated reduction in quantity with each cell division, mitochondria and chloroplasts need to be able to replicate themselves. These organelles contain their own small amounts of DNA and replicate independently of the nucleus, in this way maintaining their numbers in successive generations of cells. By the time the daughter cells have grown to the size of the original cell, organelle replication has occurred to restore the organelle number to that of the original cell. **Assimilation** is important in the growth of many organelles, such as the endoplasmic reticulum.

The DNA in mitochondria is referred to as mtDNA (mitochondrial DNA). Molecules of mtDNA are much shorter than nuclear DNA molecules and are arranged in a single circle.

You will learn more about the structure of mtDNA, maternal inheritance and applications of mtDNA sequencing in Chapter 4.

In gamete production in humans, all mitochondria inherited by offspring are located in the cytoplasm of the egg cell. Sperm cells are tiny and have very little cytoplasm, because they are made up predominantly of a nucleus and a tail for swimming. Therefore children inherit all their mtDNA from their mother. No mtDNA is inherited from the father. Therefore, studies of mtDNA reflect maternal inheritance over many generations.

KEY CONCEPTS

- There are two types of nuclear division – mitosis and meiosis.
- The repeating sequence of cell growth and division that a cell passes through is called the cell cycle, with phases G_1, S, G_2 and M.
- Specialised cells are differentiated and no longer go through the cell cycle.
- Mitosis has five phases: interphase, prophase, metaphase, anaphase and telophase.
- Interphase is G_1, S and G_2 of the cell cycle. DNA replication takes place during the S phase.
- In metaphase, sister chromatids held together by a centromere line up on the equator of the cell.
- In anaphase, the sister chromatids separate from each other and move towards opposite poles, drawn by contraction of the spindle fibres.
- Cytokinesis is division of the cytoplasm and begins as nuclear division is ending.
- Cells that form by mitosis have the same genetic material as their parent cell and each other.
- Organelles in the cytoplasm that have their own DNA, such as mitochondria and chloroplasts, replicate independently of the nucleus.
- Telomeres decrease in length with each division, and short telomeres are a sign of ageing.

CHECK YOUR UNDERSTANDING 3.1

1 Define each of the terms below and draw a fully labelled diagram to distinguish between them: chromatin, chromosome, chromatid, centromere.

2 Explain the difference between a *chromatid* and a *chromosome*.

3 Distinguish between *cell division* and *mitosis*.

4 **a** Describe the main difference between unicellular organisms and multicellular organisms with respect to the role of mitotic cell division.

b State two other important roles of mitosis in multicellular organisms.

5 Compare cytokinesis in plant cells with that in animal cells.

a Draw a fully labelled comparative diagram, to compare early prophase with metaphase in a cell that is dividing by mitosis.

b Explain why more cells are found in interphase than in any other phase of mitosis in the field of view of the dividing region of a root tip cell.

3.2 DNA structure – the Watson and Crick model

Today we know that cells carry their biological information in a molecule called DNA (deoxyribonucleic acid). However, the actual chemical nature and chemical structure of the hereditary material and genes was a mystery until the middle of the 20th century.

A brief history of the discovery of DNA

In 1869, Swiss scientist Friedrich Miescher discovered a chemical he called 'nuclein', and showed that this distinct chemical was commonly found in the nuclei of cells in a variety of tissues. His 'nuclein' proved to be made up of DNA and associated proteins from cell nuclei. Miescher is therefore credited with being the first scientist to identify DNA as a distinct molecule.

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By 1910, chromosomes had been discovered. At that stage, scientists knew that they were made up of a mixture of DNA and protein and that they carried the units of heredity called genes (named in 1909 by William Johannsen).

Over the next 30 years or so, the common expectation among scientists was that the secret of heredity would be found in proteins, because their building blocks were more varied – twenty amino acids, compared with DNA's four bases. However, by the mid-20th century, a number of scientists were moving towards the idea that DNA carried the genetic information.

In 1944, Oswald Avery and his team, working on bacteria at the Rockefeller University hospital in New York, first provided experimental proof that DNA, not protein, is the hereditary material.

Many biologists were not convinced, as they were suspicious of Avery's methodology and thought he may have obtained his results using DNA that had been 'contaminated' by protein. The race was on in laboratories around the world to try to solve the puzzle – the structures of both protein and DNA were being investigated.

Two leading teams in England were working on the molecular structure of biological molecules at the time:

- James Watson and Frances Crick worked in the Cavendish laboratory at Cambridge University, under the leadership of Lawrence Bragg.
- Maurice Wilkins and Rosalind Franklin worked at King's College in London, under the leadership of John Randall.

Worksheet
The discovery of DNA

By 1951, highly competitive teams of scientists in these British laboratories were doing similar research, as was the American laboratory of Linus Pauling. There was a longstanding rivalry between Lawrence Bragg and Linus Pauling (who had won a Nobel prize for his work on chemical bonds). Early in 1953, Linus Pauling proposed that DNA was the heredity molecule and that its structure was a *triple* helix.

Watson and Crick used chemical and X-ray evidence to construct a model of the molecular structure of DNA (Fig. 3.9). The model revealed a stable molecule that could store large amounts of information. It was a two-stranded molecule, with paired bases twisted into a spiral ladder. Because of its structure, they called the DNA molecule a 'double helix' (and the name of the book that Watson later wrote, describing their journey of discovery, was *The Double Helix*). An interesting and critical feature of their model was that it could be used to explain how the molecule could self-replicate.

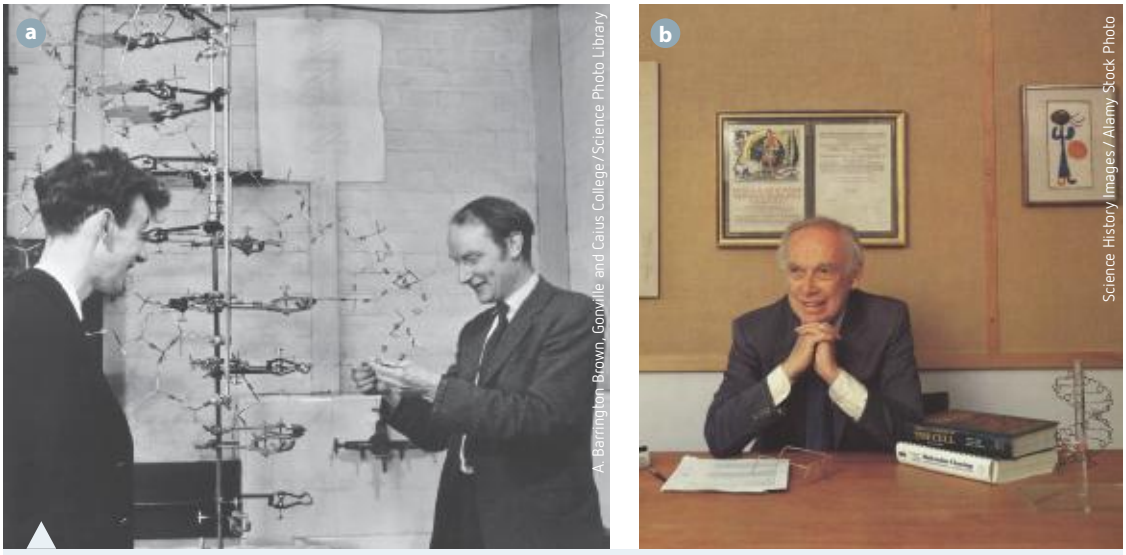

FIGURE 3.9 **a** James Watson (left) and Francis Crick (right) with their model of DNA structure (1953); **b** James Watson in later years, with some of the many books he wrote about DNA and science

Watson and Crick's model of DNA showed a large linear molecule arranged as a double helix or 'twisted ladder', with four nitrogenous bases that were held in pairs by hydrogen bonds. Their model suggested that genetic information could be stored in the order or sequence of the bases that make up the middle of the DNA structure. On 28 February 1953, Watson and Crick won the race, and announced that afternoon, at a pub called The Eagle, that they had discovered 'the secret of life'.

FIGURE 3.10 **a** The Eagle pub in Cambridge, England, and **b** the plaque outside the pub to commemorate where Crick and Watson celebrated their discovery

Weblink
TED talk: James Watson speaking about how he discovered DNA

Weblink
Discovery of the structure of DNA
The full story of the scientific advances preceding Watson and Crick's insightful discovery

Watson and Crick had worked collaboratively, evaluating the processes, claims and conclusions of other scientists. They had asked questions of colleagues such as Maurice Wilkins who, in response, had shown them Rosalind Franklin's X-ray crystallography photograph of DNA (without first asking her permission) to Watson when he visited the laboratory. Watson and Crick applied critical and creative thinking, both in predicting what to expect and in analysing the primary and secondary chemistry and crystallography data they had gathered.

They considered the quality of all available evidence from a variety of scientists and used reasoning to construct their scientific predictions and arguments, subjecting the model they were building to rigorous testing and validation. As a result of their innovation, their attention to sound scientific detail and their collaborative approach, they came to the valid conclusion that won them a Nobel Prize in 1962. Avery and his team may also have been in line for a Nobel Prize but, like Rosalind Franklin, Avery died before this prize could be awarded to him.

DNA structure – the Watson and Crick model

Crick realised that each DNA strand, comprising a sugar-phosphate backbone, is antiparallel to the other, meaning that the twisting strands run parallel to each other but in opposite directions. X-ray crystallography photographs of the DNA structure suggested a set distance between the 'backbones' of the molecule. Thus, each base could have one end attached to the backbone and the other end hydrogen-bonded to another base. So the bases lined up between the backbones, like the rungs of a ladder (Fig 3.11).

Nucleotide pairing and bonding

Watson and Crick realised that in their DNA model, to ensure the DNA backbones remain equidistant from each other, a double-ringed purine base needs to bond with a single-ringed pyrimidine base. The purine base adenine must therefore bond with the pyrimidine base thymine (A and T), and the purine base guanine must bond with the pyrimidine base cytosine (G and C).

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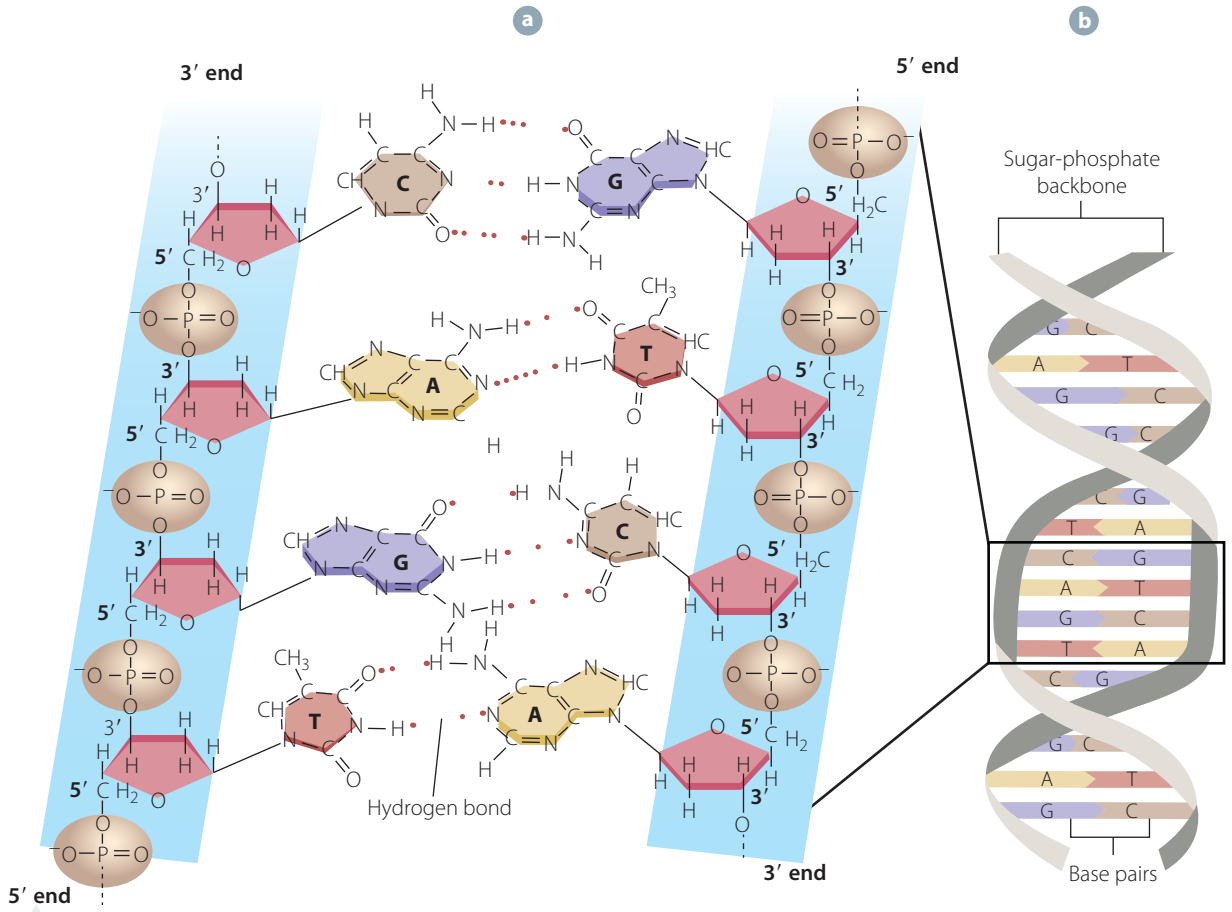


FIGURE 3.11 The chemical structure of DNA, showing its antiparallel backbone and base pairing: **a** the molecular structure of DNA showing the sugar-phosphate bonding and hydrogen bonding of base pairs; **b** a simplified version of DNA

From the chemistry of this pairing, they also predicted that the DNA strands would be held together by weak hydrogen bonds between the paired bases: a double bond between A and T, and a triple bond between G and C (Fig. 3.12). Watson and Crick found evidence for this pairing, as it fitted with Chargaff's 'parity rule'. In 1951, Austrian biochemist Erwin Chargaff discovered that, in DNA, the ratio of the bases A:T and C:G was always 1:1.

Watson and Crick realised that this complementary base pairing could also be used to predict how DNA could be copied. They published their research paper, titled, 'Molecular structure of nucleic acids', in 1953. In this paper they stated: 'It has not escaped our notice that the specific pairing we have postulated immediately suggests a possible copying mechanism for the genetic material'.

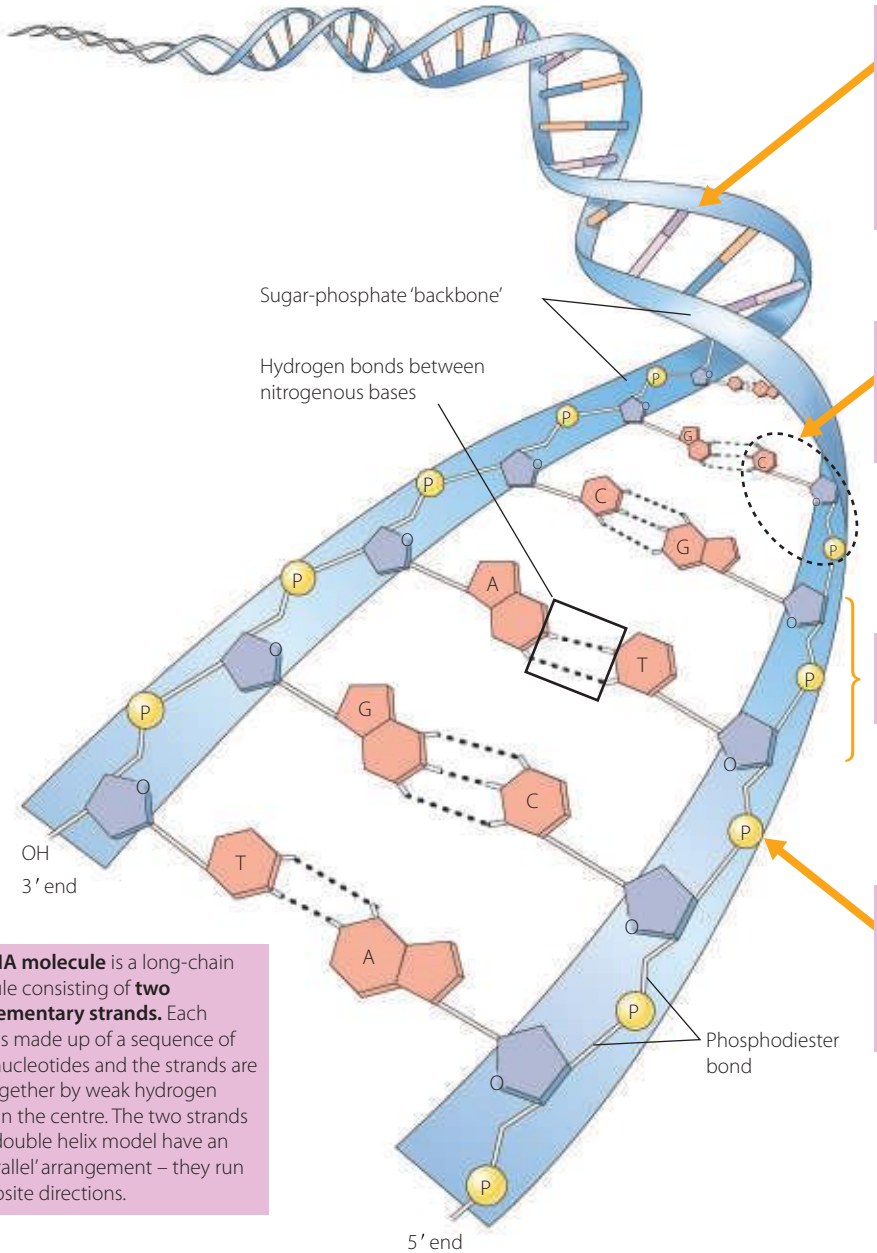
DNA serves as a template for replication

Watson and Crick's DNA model showed all the requirements expected of hereditary material.

- DNA can carry, in coded form, all the instructions for the formation and functioning of cells, despite the fact that its 'alphabet' consists of only four nitrogenous bases.
- The structure of DNA allows for its own replication. To copy DNA, each strand can serve as a template for enzymes to synthesise a new complementary DNA strand.

Weblink
The double helix
Watch this 16-minute film about the discovery of DNA.

DNA can be transferred from one generation to the next, packaged in the form of chromosomes and carried by gametes. It was discovered that proteins associated with DNA in chromosomes have the role of keeping DNA 'neatly packaged'. DNA is coiled around these proteins (called **histones**), in much the same way that cotton thread is wound around a cotton reel. This holds the long threads of DNA in a compact way so that DNA is easy to sort and separate during cell division and can be efficiently transported from one generation to the next. Histones are also thought to play a role in exposing sections of DNA, so that genes can be expressed.



The vertical sides of the ladder are made up of alternating sugar and phosphate molecules (a **sugar-phosphate backbone**) and the 'rungs' of the ladder are pairs of nitrogenous bases (adenine, thymine, guanine and cytosine or A, T, C and G).

Each **nucleotide** is made up of a phosphate group, a sugar (deoxyribose sugar), and a nitrogenous base attached to the sugar.

There are four types of **nitrogenous bases:** adenine, guanine, cytosine and thymine.

Chemically, **bases pair** in a particular manner: adenine with thymine and guanine with cytosine (i.e. **A–T** and **G–C**). They are held together in the centre by **hydrogen bonds,** forming two complementary strands.

The **DNA molecule** is a long-chain molecule consisting of **two complementary strands.** Each strand is made up of a sequence of many nucleotides and the strands are held together by weak hydrogen bonds in the centre. The two strands in the double helix model have an 'antiparallel' arrangement – they run in opposite directions.

FIGURE 3.12 An annotated diagram of the DNA double helix molecule, showing sugar-phosphate backbone and base pairing

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TABLE 3.3 Validation of the model: Watson and Crick's findings, and the evidence on which they were based

FINDING	EVIDENCE-BASED REASONING
The whole DNA 'ladder' molecule, instead of being flat, spirals and is therefore known as the 'double helix'.	*Evidence*: X-ray crystallography suggested a helix measuring 3.4 nm for every turn, and this fitted the model where exactly 10 base pairs would measure 3.4 nm in length and make up one twist of the helix.
The four types of nitrogenous bases: adenine (A), guanine (G), cytosine (C) and thymine (T) always pair as A–T and C–G.	*Evidence*: Chargaff's rule stated that DNA from any cell has a 1:1 ratio of pyrimidine and purine bases and, more specifically, that the amount of guanine is equal to cytosine and the amount of adenine is equal to thymine. Watson and Crick found complementary base pairing of pyrimidine–purine (that is, A–T and G–C) was critical in keeping the backbones of DNA equidistant.
The two complementary strands of DNA are held together in the centre by hydrogen bonds that form between the complementary bases.	*Reasoning*: Chemically, when A bonds to T, a double hydrogen bond may form and when G bonds to C, a triple hydrogen bond may form.
The strands of the backbone must be identical and run in opposite directions (antiparallel).	*Evidence*: DNA crystal images, generated by Rosalind Franklin, looked the same when they were turned upside down or backwards. *Reasoning*: The backbones are made of sugar-phosphate molecules, based on the ratio of these in the chemical analyses.
Each DNA strand serves as a template for the production of a complementary strand, allowing the double-stranded molecule to self-replicate.	*Reasoning*: The two complementary strands of DNA could 'unzip' or open up if the hydrogen bonds break between the base pairs, allowing them to replicate.
Watson and Crick could form hypotheses about DNA's structure by building physical models of how the atoms fit together. Sometimes they used cardboard cut-outs to represent the four nitrogenous bases and other subunits of DNA. They played with these on a desk, like a jigsaw puzzle. They initially made an error in the configuration of the rings in thymine and guanine. By changing the arrangement of atoms and making new cut-outs, based on a suggestion from Jerry Donahue, an American scientist, they found the perfect fit: complementary base pairing, ratios to reflect Chargaff's rule and the hydrogen bonding of purines to pyrimidine bases.	

Nucleic acids as we know them today

There are two types of nucleic acids – DNA (deoxyribonucleic acid) and RNA (ribonucleic acid). Nucleic acids contain the chemical elements carbon, hydrogen, oxygen, nitrogen and phosphorous.

All nucleic acids are polymers made up of simple repeating units (monomers) called **nucleotides**. Nucleotides may be linked together to form single chains, as in RNA, or double strands, as in DNA (Fig. 3.13). These DNA strands may be very long. For example, human chromosome 1 contains 249 million base pairs of DNA, representing approximately 8 per cent of the total DNA content of a human cell.

Each nucleotide is made up of three parts: a simple sugar (ribose in RNA, deoxyribose in DNA), a phosphate and a nitrogenous base.

The genetic code is created in the consecutive sequence of bases. These base sequences differ in each gene, providing the 'genetic code' for a cell.

Worksheet
Chromosomes – the key to inheritance

Comparing the roles of DNA and RNA in cells

- DNA stores the genetic information that controls the cell and thereby the whole organism.
- DNA is the main chemical making up chromatin in the nucleus, although small amounts of DNA are also found in mitochondria and chloroplasts.
- DNA is responsible for transmitting inherited information from one cell to another during cell division and from one generation to another during reproduction.
- RNA is a nucleic acid found in small amounts in the nucleus, and in larger amounts in the cytoplasm; it is usually associated with ribosomes.
- RNA has the base uracil (U) instead of thymine (T).

Revise the structure of nucleic acids from *Biology in Focus Year 11*, Chapter 3

Worksheet
The structure of nucleic acids

The three types of RNA and their functions are dealt with in more detail in Chapter 4. See Fig. 4.12.

DNA instructing the formation of proteins is also dealt with in Chapter 4.

- There are three types of RNA:
  - messenger RNA (mRNA), which is involved in carrying information from DNA, transporting a transcribed copy from the nucleus to the cytoplasm
  - ribosomal RNA (rRNA), which is associated with proteins in ribosomes, bringing mRNA and tRNA together during translation
  - transfer RNA (tRNA), which assists in 'translating' the mRNA message into proteins. (You will learn more about this in the next section.)

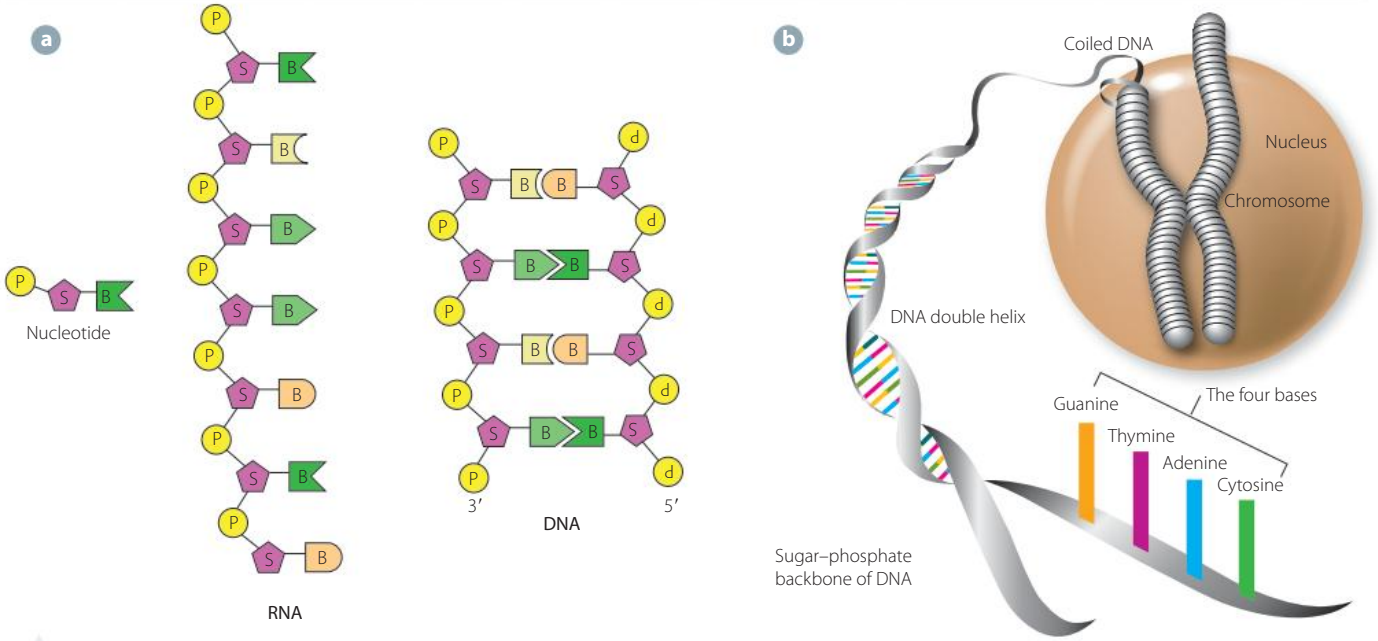

FIGURE 3.13 **a** The nucleotide monomer is made up of base–sugar–phosphate, a single-stranded RNA polymer molecule or polynucleotide, and a double-stranded DNA molecule (a polymer of nucleotides). **b** The four bases and sugar-phosphate backbone make up the DNA, which forms a chromosome. Here the chromosome is composed of two identical (sister) chromatids, held together by the centromere.

DNA instructs the formation of proteins

See page 125 for the genetic codes for amino acids.

In 1955, Crick proposed that the order of the four bases (A, T, G, C) on the DNA molecule determines the sequence of amino acids in a protein. Their 'sequence hypothesis' could explain how the order of bases in genes along the chromosome can be translated into the language of proteins. By 1966, scientists had cracked the genetic code and could show exactly which three-base sequences code for each amino acid.

KEY CONCEPTS

- Each nucleotide consists of three parts – a phosphate, a sugar (deoxyribose sugar in DNA, ribose sugar in RNA) and a nitrogenous base.
- There are four types of nitrogenous bases and each nucleotide is named after the base that it carries – adenine, thymine, guanine or cytosine nucleotides. These are often simply referred to by their first letters – A, T, G and C.
- A and G are the larger double-ringed purine bases, and C and T are the smaller single-ringed pyrimidine bases.
- Each DNA molecule is made up of two chains or strands that have an antiparallel arrangement – that is, they are parallel but run in opposite directions.
- Each strand is made up of a sequence of many nucleotides, and the strands are held together by weak hydrogen bonds between the bases in the centre of the DNA molecule.
- The advantage of these weak hydrogen bonds is that little effort is required to pull the bases apart so that DNA can replicate or be decoded to form proteins.

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- The vertical sides of the ladder are made up of alternating sugar and phosphate molecules (a sugar-phosphate backbone) and the 'rungs' of the ladder are pairs of a purine base linked to a pyrimidine base: adenine-thymine (A-T) and guanine-cytosine (G-C). The bases are attached to the backbone through the sugar component.
- The bases are arranged in a sequence along each strand of DNA (e.g. GGTCAGGCTTGAACGA) and the length of a DNA molecule is presented as the number of base pairs (bp).
- The two strands of DNA unzip (weak hydrogen bonds break) for replication and gene expression.
- RNA is a single-stranded molecule made of nucleotides, with a ribose sugar attached to each phosphate to form the backbone; on the other end it attaches to a base, either A, U, C or G. RNA does not have thymine, which is replaced by the base, uracil (U).

CHECK YOUR UNDERSTANDING 3.2

1. Create a timeline to summarise the contributions made by scientists in the discovery of the hereditary material in cells. List the scientists in the chronological order of their discoveries and summarise their findings under headings such as: Date, Name(s), Laboratory/Place of work, Contribution (theory/law/hypothesis), and a brief description of findings in their research.
2. Describe the main features of DNA that Watson and Crick discovered, under the headings: Nucleotide composition, Base pairing and Bonding.
3. Compare, using examples, the structure of a purine with a pyrimidine base.
4. Explain, in terms of the structure of DNA, why a purine had to combine with a pyrimidine to create a double helix.
5. What is meant by the term 'antiparallel' in describing the backbones of DNA?
6. Outline the three main requirements of hereditary material for it to be able to carry out its functions.
7. How does the structure of DNA support the idea that it can self-replicate?
8. Compare the structure and functions of DNA and RNA.
9. Evaluate the benefits and limitations of the two DNA models shown in Fig. 3.11.

3.3 DNA replication – the Watson and Crick model

DNA replication is the production of two identical double-stranded molecules of DNA from one original double helix molecule. DNA replicates during interphase prior to mitosis, and each cell receives one exact copy of the coded instructions that control the basic life functions of the cell.

Watson and Crick noted that their model suggested a possible copying mechanism for the genetic material. If the strands separated, each strand could act as a template for the synthesis of a new, complementary strand. This is indeed what happens. This process of DNA replication in cells is termed 'semi-conservative', as each resulting double-stranded DNA molecule has one new strand and one 'old' or conserved strand. This mechanism should ensure that the genetic material is copied exactly every time.

However, DNA replication is not quite as simple as it appears. The following features add complexity to the process:

- DNA has to be unwound from its spiral configuration before the strands can be separated.
- A large number of physical and chemical reactions take place simultaneously during DNA replication, and so a collection of enzymes is required to control each reaction.
- The two strands of DNA run in opposite directions (antiparallel) and nucleotides can be added in one direction only (onto the free 3′ end).
- DNA replication errors sometimes occur and these need to be corrected.

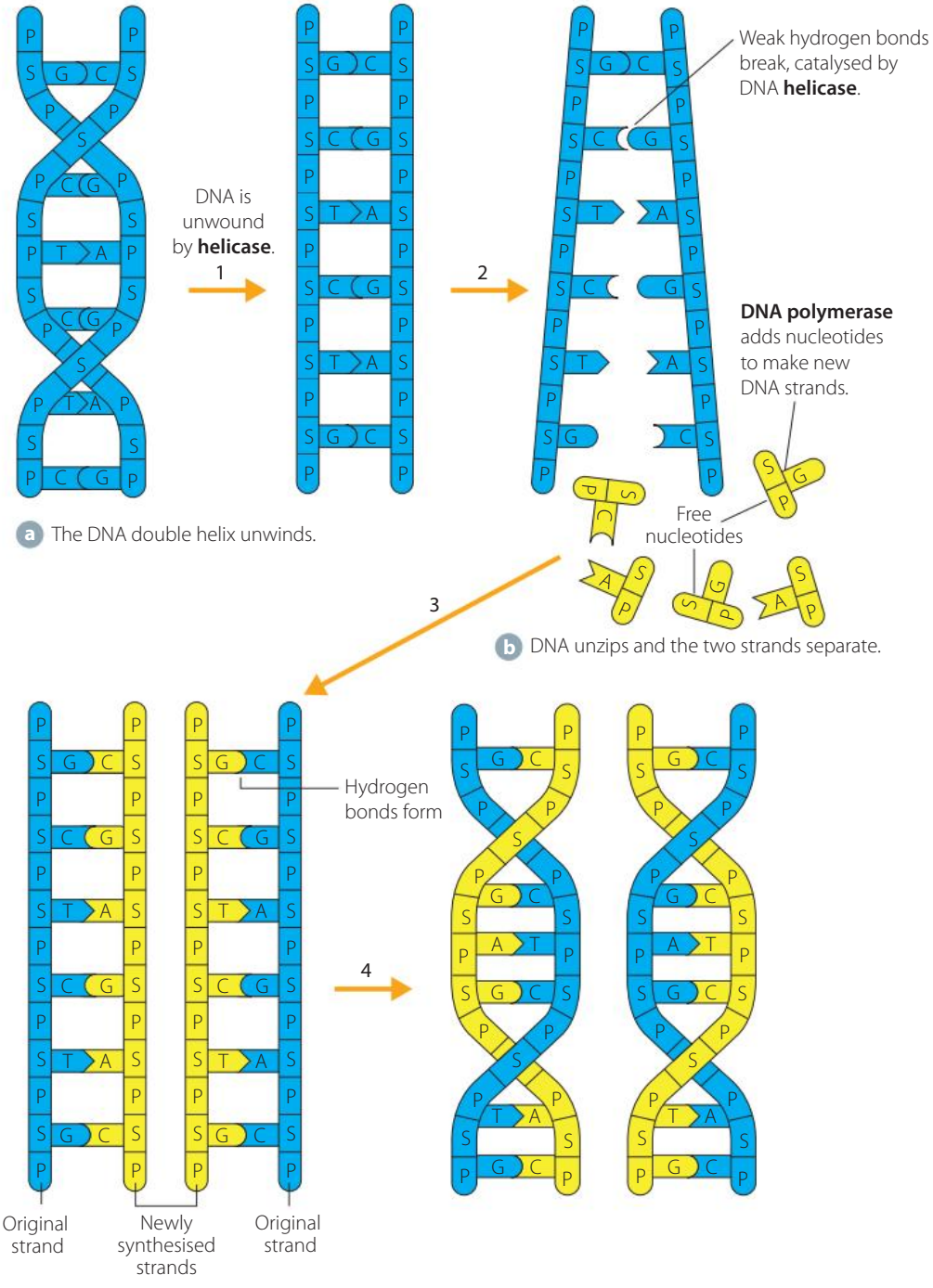
The process of DNA replication

DNA replication occurs in four main steps.

1 *The DNA double helix unwinds.*

Each DNA molecule is a double-stranded helix. An enzyme called **helicase** causes the DNA helix to progressively unwind and the strands to separate (Fig 3.14a).

Worksheet
DNA replication



a The DNA double helix unwinds.

b DNA unzips and the two strands separate.

c Nucleotides are added alongside both strands, opposite their complementary bases, to create a new strand along each original strand.

d The resulting DNA molecules each contain half of the original DNA molecule and a newly synthesised strand (semi-conservative replication).

FIGURE 3.14 DNA replication – a simplified diagrammatic sequence

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2 *DNA unzips – the two strands separate.*

Using ATP as the energy source, helicase disrupts the weak hydrogen bonds between the complementary bases of the nucleotides on opposing strands. The DNA strands separate, exposing the nucleotide bases (Fig. 3.14b). If you think of DNA as a 'ladder', each 'rung' splits down the middle, starting at the bottom of the long DNA molecule and creating a **replication fork** further up, where the DNA is still joined (Fig 3.15). Single-stranded binding proteins (SSBs) bind to and stabilise the newly separated single-stranded DNA.

3 *Nucleotides are added against each single strand.*

Each separate strand of the DNA molecule acts as a template for the production of a new strand of DNA. For synthesis to be initiated, a short strand of RNA needs to be made and attaches to the DNA – this is known as a **primer**. RNA primers are made by the enzyme **primase**.

Next, the enzyme **DNA polymerase III** adds DNA nucleotides, to continue the synthesis of the new strand. It picks up free nucleotide units (made of sugar–phosphate–base) floating in the liquid background of the nucleus and inserts them opposite their complementary base partner, using the existing strands as templates and keeping the nucleotide pairing specific:

Adenine (A) always pairs with thymine (T) nucleotides.
Guanine (G) always pairs with cytosine (C) nucleotides.

Note: The two strands in a double-stranded DNA molecule run in opposite directions – they are antiparallel. Each DNA strand has a three prime (3′) end and a five prime (5′) end. Nucleotides are always added from the three prime (3′) end of a DNA strand. Therefore nucleotide insertion during replication is also antiparallel along the two template strands.

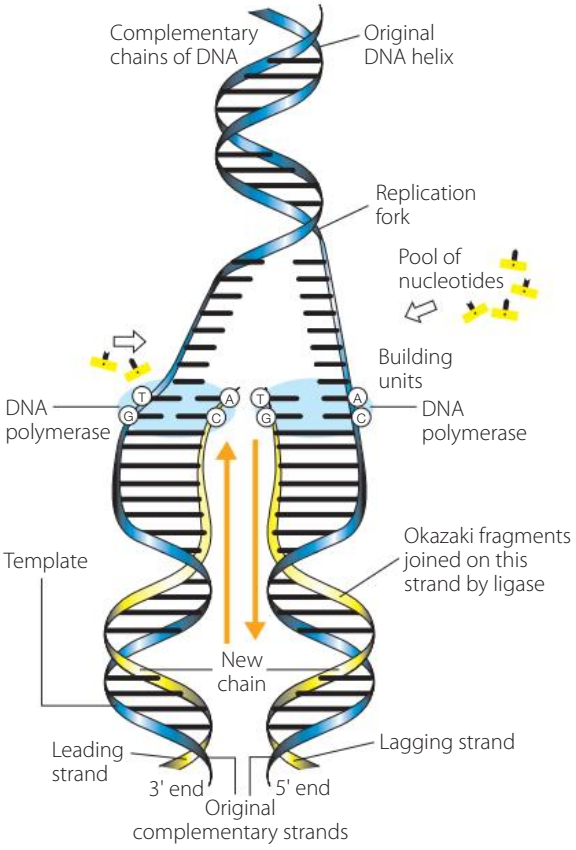


FIGURE 3.15 DNA replication showing a replication fork, free nucleotides and DNA polymerase synthesising the new complementary strands (drawn in yellow)

On one strand of DNA, nucleotides are added in a long chain, growing in the same direction as the replication fork opens up. This is called the **leading strand** and replication is continuous.

On the other strand (the **lagging strand**), nucleotides are added in 'chunks' (called Okazaki fragments), from the replication fork backwards (Fig. 3.15). Replication in this strand is therefore discontinuous and the fragments are then joined up by an enzyme called ligase, to form one continuous strand.

Base mismatch pairs are dealt with in more detail on page 104 (Fig. 3.18).

4 *Replication errors are identified and corrected.*

DNA polymerase I is a complex enzyme that can backtrack to 'proof read' and 'edit' the strand. It corrects any base pair errors by splicing out the incorrect base that was inserted into the new strand and replacing it with the correct base.

Finally, the two new strands are sealed together by the enzyme called **ligase**. The final base pairing is checked by another DNA polymerase enzyme, which recognises base pairing errors and carries out base mismatch repairs, to ensure accuracy.

Despite these checking mechanisms, a small number of errors in DNA replication still occur – about one in every ten billion base pairs. Incorrect base pairing results in a change in the DNA base sequence, known as a mutation.

Cells may delay the progression of the cell cycle until DNA repair (during the G_2 phase) is complete.

Each resulting DNA molecule contains one strand of the existing DNA molecule and a newly synthesised strand. The replicated DNA molecules rewind into the double helix conformation, like

Weblink
DNA replication
Watch this review of DNA replication

the original molecule. The end result is that there are two molecules of DNA, each a double-stranded helix, identical to each other and to the original molecule from which they formed.

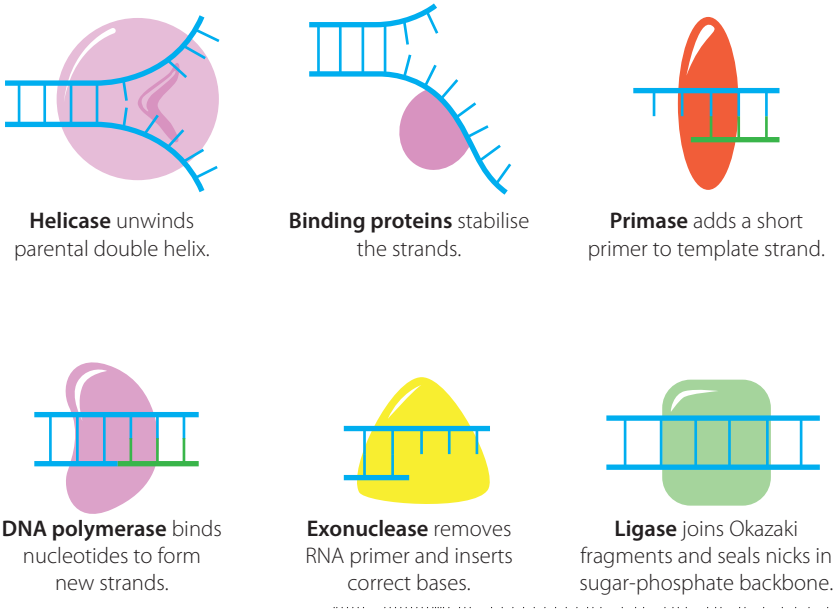
Enzymes ensure exact replication of DNA

Replication of DNA is a complex process and each step is controlled by one or more enzymes (Table 3.4, Fig. 3.16). Most spontaneous changes in DNA are temporary because they are immediately corrected by repair enzymes.

TABLE 3.4 Enzymes that regulate DNA replication

ENZYME	FUNCTION IN DNA REPLICATION
Topoisomerase (e.g. gyrase)	• Relaxes DNA from its supercoiled state, always working ahead of the replication fork
Helicase	• Follows topoisomerase and unwinds the double helix by breaking hydrogen bonds between bases, causing the two strands to separate and creating a replication fork
Primase	• Connects RNA primer to a strand to initiate DNA replication • Synthesises a short complementary RNA molecule, which binds to DNA, serving as the starting point for DNA synthesis by polymerase
DNA polymerase III	• Synthesises new DNA strands, using existing strands as a template. Nucleotides are added to a growing strand from the 3′ end • Joins the phosphate group of each nucleotide to the previous one, creating phosphodiester bonds between these molecules to make the sugar-phosphate backbone
DNA polymerase I	• Mainly functions in 'editing' – recognises and repairs base pairing errors (exonuclease) • Also has a function in replication (removing primers ahead of the main polymerising enzyme)
DNA polymerase II	• Editing function, but no exonuclease activity
Ligase	• Connects and seals the two strands of the DNA molecule and also connects Okazaki fragments
• On the leading strand – DNA moves along in the same direction as the developing replication fork, and nucleotides are added in one long chain. • On the lagging strand, DNA is added one 'chunk' at a time – called Okazaki fragments (about 100–150 nucleotides long) and these are then joined up.	

FIGURE 3.16 Enzymes involved in DNA replication



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INVESTIGATION 3.2

Modelling DNA replication using the Watson and Crick model

BACKGROUND: USING SCIENTIFIC MODELS AND WRITING A HYPOTHESIS

Information and communication technology capability

Critical and creative thinking

Following their landmark paper on DNA structure, in 1954 Watson and Crick proposed a mechanism by which DNA could be copied. This mechanism was based on their DNA model in that each DNA strand could serve as a template for the synthesis of a new strand. In their own words, 'prior to duplication, the hydrogen bonds are broken, and the two chains unwind and separate. Each chain then serves as a template for the formation … of two pairs of chains, where we only had one before. Moreover, the sequence of the pairs of bases will have been duplicated exactly.' So the final result is two identical double helix DNA molecules (sister chromatids), each made up of one old and one newly synthesised strand – a model of semi-conservative replication.

PREDICTIONS AND VALIDATIONS OF THE WATSON AND CRICK MODEL OF SEMI-CONSERVATIVE REPLICATION

Weblink
Hypotheses used to test DNA replication proposal
Investigative methods used to test the hypothesis that DNA replication is semi-conservative

The semi-conservative model that Watson and Crick proposed was tested and validated by scientists using the DNA of both prokaryotes and eukaryotes.

Scientists realised that, to test the Watson-Crick proposal for DNA replication, they needed to find a way to distinguish between the old and the newly synthesised strands.

In 1958, Matthew Meselson and Franklin Stahl carried out experiments that supported and verified Watson and Crick's proposal of semi-conservative replication of DNA. They used heavy isotopes of nitrogen, which were incorporated into the nitrogenous bases that would be added to the newly synthesised strand. A centrifuging technique was then used to determine the density of the DNA strands, determined by the buoyancy of DNA. The results of the buoyancy tests supported predictions made for semi-conservative replication.

VALIDATING SCIENTIFIC MODELS

Scientific models are developed to represent an idea, object, process or system that cannot be observed directly. Models are simplified representations; they do not try to explain every detail in the 'real world' process, object or idea. Scientists are usually aware of the limitations of the models that they propose and have critically analysed. The models that are most valuable in science are those that allow us to make and test predictions. If a prediction is made and the model holds true, the prediction is said to validate the model. Sometimes, as new evidence is found, models need to change to reflect a more accurate understanding of an idea or process. If a model cannot hold true in the light of new evidence, the model may be rejected. Models are therefore indicative of the tentative nature of science.

Weblink
Cut and paste DNA

AIM

To model DNA replication, showing semi-conservative replication

In this investigation, you are required to generate a model to represent the structure of DNA and the process of DNA replication. Because each model created will demonstrate only some aspects of the structure and process, the models built by students will all be different. You will be required to peer assess the models that you view. You will need to identify the structural and conceptual differences between the models and use your critical analysis skills to discuss the advantages and limitations of each model.

Weblink
DNA with confectionary

METHOD

1. Working in groups of three to four, build a working model of DNA structure, showing the double helical nature of the molecule. You need to build a working model with features and spare parts that will be used by another group to demonstrate how your DNA can act as a template to demonstrate semi-conservative replication. Your teacher may assign one or two specific features (a to e below) that your model needs to show, or each group may be allowed to select which features they wish to show.
2. Using the Internet, carry out research to review different types of DNA models that may be created to show structure and/or replication. You will discover that models may be created from a wide variety of materials, from edible models using lollies, to models that use materials such as pipe cleaners and beads, or easily available recyclable materials such as cardboard and toothpicks, or waste materials such as styrofoam and plastic. See the online resources linked to this page for ideas. You will also need to analyse models of DNA

Weblink
Digital DNA

»

» replication to determine whether they can be used to demonstrate the feature of replication you have selected or been assigned.

3 Your model of semi-conservative replication of DNA should demonstrate some or all of the following:
   - **a** complementary pairing of bases
   - **b** creation of Okazaki fragments
   - **c** unwinding of DNA with helicase action, creating a replication fork
   - **d** addition of nucleotides by DNA polymerase, using an old strand as a template and creating a new strand
   - **e** mutation during DNA replication.

4 Label the structures in the DNA model. Labels should include all the features listed in Table 3.5. In addition to these labels, also label any additional features, such as enzymes or assigned features.

   Common labels: sugar-phosphate backbone, nucleotide, nitrogenous base, hydrogen bond, 3′ end, 5′ end.

PREPARING YOUR MODEL FOR VALIDATION

5 Create the following to be placed with your model in the next lesson:
   - **a** instructions to your classmates on how to replicate your DNA molecule
   - **b** raw materials, so that another group can carry out the replication process on your model
   - **c** a copy of Table 3.5, completed to describe the features of your model
   - **d** a printout of a photograph of your model (before it is replicated).

TABLE 3.5 Key to DNA model

FEATURE	MATERIAL	COLOUR
Sugar-phosphate backbone		
Nitrogenous bases		
Hydrogen bonds		
One nucleotide		
Feature to be demonstrated during replication		
Limitations		

GROUP ROTATION ACTIVITY: VALIDATING MODELS

6 Set up your model and provide spare parts and clear instructions for the next group on how to replicate your DNA molecule. Copy and complete Table 3.5 to accompany your model as a key, identifying what materials have been used to represent each feature. You also need to provide a printout of a photo of your model.

7 Rotate groups: A → B; B → C; C → D; D → E; E → A.
   - **a** Identify the parts of the DNA model you have rotated to, using the key provided. Take a photo of the model before you replicate it.
   - **b** Use the spare parts provided to carry out replication, showing the main feature listed at the bottom of the table. You may use your textbook to revise exactly what this feature is. Take a photo of the replicated model. Complete a peer assessment of the model, listing its advantages and limitations.

8 Move to the next group and take on the proof reading and editing role of DNA polymerase. Check whether the model has any errors of replication (mutations). Take a photo of the replicated model. Turn the explanatory table over (or cover it with a piece of paper) so that the next group cannot see it.

9 Move to the next group and complete a blank copy of Table 3.5 for the model you are viewing. Do not cheat by turning over the key. Take a photo of the model and your completed table.

10 Move to the last model. Compare it with the model that your group made, writing down two similarities and one difference between this model and your model. Take a photo of the model.

Weblink
DNA replication
Build strands of DNA by inserting complementary bases

EXTENSION

DNA replication – try the DNA replication simulation in class or complete it for homework. (See the weblink.)

KEY CONCEPTS

- The two DNA strands unzip for replication.
- DNA polymerases are complex enzymes that move along the DNA molecule, linking nucleotides to their complementary base partners to make new DNA chains.
- Each strand of DNA is used as a template to make a new, complementary strand – this is known as semi-conservative DNA synthesis.
- Accurate DNA replication is important so that daughter cells have exact copies of the genetic information for synthesising proteins.
- Some DNA polymerase enzymes have the ability to 'proof read and edit' the DNA, correcting its own errors. There are also complex enzymes to repair other damage to DNA caused by mutagens.

CHECK YOUR UNDERSTANDING 3.3

1 What is meant by 'DNA replication'?
2 Create a flow chart that accurately shows the steps in the process of DNA replication. Indicate any enzymes that act at particular steps and what their function is.
3 What is an error in the DNA base pairing called and how does it arise?
4 Why is DNA replication called 'semi-conservative'?

3.4 The importance of accuracy during DNA replication

Because the sequence of bases in DNA makes up the genetic code of an individual, exact copying of this sequence during replication is critical, for two main reasons:

- heredity (inheritance of genes) – the genetic material transmitted from cell to cell (by mitosis) and from generation to generation (by gametes from meiosis) needs to be accurate
- gene expression (protein synthesis) – the genetic instructions given to a cell to create its structure and ensure its correct functioning must be accurate.

It is therefore very important that no errors are made when this information is replicated. This is termed fidelity of replication.

However, studies show that DNA is at constant risk of mutation. It is therefore not surprising to find that cells have enzymes that are able to repair incorrect base insertions and other DNA damage that may arise during replication.

Causes of mutation and types of mutation are dealt with in more detail in Chapter 7.

Errors in replication

Natural errors that arise at random during DNA during replication are called spontaneous mutations. Other errors that arise as a result of exposure of cells to environmental factors such as radiation or chemicals are called mutagenic mutations. Environmental factors such as radiation, chemicals and viruses that change DNA are called mutagens. As the length of time that the cells are exposed to mutagens increases, and the intensity of exposure rises, so the risk of mutation also increases.

There are enzymes in cells to repair both types of mutation, but sometimes DNA errors go undetected and this results in a permanent mutation. This uncorrected mutation will be replicated in successive divisions and, if occurring in meiosis, passed on to later generations of individuals.

FIGURE 3.17 An uncorrected mutation in DNA will be passed on to future generations.

DNA repair

The insertion of an incorrect base is common during DNA replication. When this occurs, a repair enzyme recognises the mismatched base pair, excises the incorrect base (cuts it out) and replaces it with the correct base. This is called DNA mismatch repair and is a function of the enzyme DNA polymerase I.

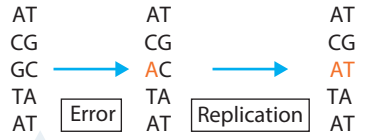


FIGURE 3.18 Mismatch error in DNA replication

However, if a mismatched pair is missed by the enzyme, harm may arise the next time the cell divides and the DNA replicates. For example, if an error occurs in which a 'C' nucleotide is accidentally paired with an 'A' nucleotide (instead of the correct 'G') (Fig. 3.18), then the next time this DNA strand acts as a template and replicates, the error will cause a strand to form that has an incorrect base pair (AT instead of GC).

This strand is now permanently mutated. The error will not be recognised by a DNA repair enzyme, because the pairing of bases is not incorrect. However, the sequence of bases within the strand now differs from that of the original strand. A permanent mutation such as this often has a harmful effect, but some mutations have a beneficial effect on the organism. Some mutations have no effect at all. It is important to remember that mutation is a mechanism that can give rise to variation in organisms and this may be beneficial to a species in terms of biodiversity and evolution.

Variation and the advantages of biodiversity in terms of evolution are dealt with in Chapter 5.

There are different types of enzymes that carry out repairs on different types of DNA damage, to ensure accurate replication and normal functioning of cells.

Gene expression and the need for accurate replication

Different types of DNA damage and their effects, as well as DNA repair are dealt with in Chapter 7.

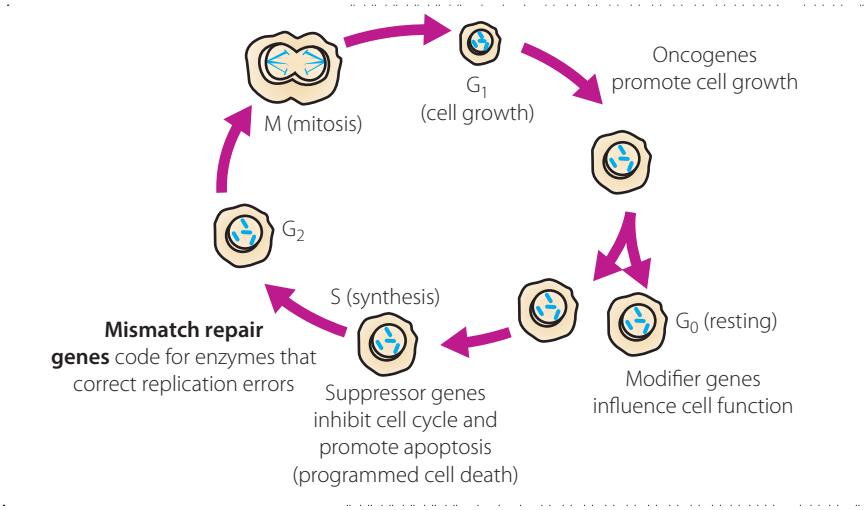
Now that you understand the structure of DNA and its ability to serve as a template during replication, you might ask how the genetic code actually works. The answer lies in the production of proteins. Enzymes are proteins; they control the synthesis of cell materials and the biochemistry (metabolism) within each cell and within the whole organism. Besides storing exact copies of instructions in every cell, the sequence of bases in DNA also plays a vital role in the translation of the genetic code into proteins.

In multicellular organisms, different genes are activated and expressed in each type of cell. The entire genetic code passed on to each cell must contain a full and accurate set of instructions, so that when the relevant gene is activated, it functions correctly to make proteins that determine the type of cell it will become. The activation of genes is regulated by other molecules, such as enzymes (which are proteins), and therefore they also need to be accurately coded for by DNA. Errors in genes that control the cell cycle may lead to changes in cell division and cell death, leading to cancer. Mismatch repair of these genes is particularly important (Fig. 3.19). Replication errors in genes that code for DNA repair enzymes are also linked to cancer.

Weblink
A comparison of two major DNA repair pathways
Analyse the information in the diagrams to understand a visual representation of DNA mismatch repair.

Genes that regulate cell division to prevent cancer are discussed in more detail in Chapter 15, pages 525–6.

FIGURE 3.19 The cell cycle, showing the stage at which mismatch repair enzymes (coded by genes) correct errors in DNA during replication



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KEY CONCEPTS

- Accurate DNA replication is important for two main reasons:
  - heredity: the genetic material of a cell must be transmitted accurately, from:
    - one cell to another during *mitosis*, allowing growth, repair and maintenance of an organism
    - one generation to another during *meiosis* (e.g. when gametes are formed for sexual reproduction)
  - gene expression: the genetic material of a cell must be transmitted accurately to give the correct instructions to a cell to ensure the correct structure, functioning and behaviour of an organism, essential for its survival.

CHECK YOUR UNDERSTANDING 3.4

1. Explain the importance of having inbuilt mechanisms for correcting mistakes in DNA replication.
2. Distinguish between a spontaneous mutation and a mutagenic mutation.
3. List three mutagens.
4. How is an error in replication, such as a mismatch repair, corrected?
5. How could an error in DNA replication end up affecting a whole organism?

3.5 The continuity of species

All living organisms arise from other living organisms. The **continuity of species** refers to the ongoing survival of species as a result of characteristics being passed from parents to offspring in a continuous lineage. This inheritance of characteristics from ancestors to currently living organisms relies on the passing on of consistently accurate genetic information (**genetic stability**) and the occasional introduction of variation of some genetic information, so that species can adapt and survive in a changing environment.

Accurate DNA replication brings about genetic stability, whereas mutation results in **genetic variation**. Although variation is important for evolution of species, genetic stability is important for the survival of the individual. Both genetic stability and variation play a role in ensuring the *continuity of the species*.

Weblink
Genetic continuity

Genetic continuity

Genetic continuity is a way of preserving genetic information across generations and is dependent on two things:

- when a cell divides by mitosis, the resulting two daughter cells must have the same number and type of genes as the original cell.
- when two sexually reproducing organisms breed, the resulting offspring must have the same number of genes as the parent organisms and variations in these genes must not be extremely detrimental or lethal.

Genetic continuity ensures continuation of a species, because it ensures that new cells or organisms have all the genes they need, in working order, to survive. A lack of genetic continuity results in disease and sometimes in death and extinction.

FIGURE 3.20 Genetic continuity across generations

Ensuring the continuity of species – genetic stability

In asexual reproduction, offspring inherit identical characteristics from one parent if replication is consistent. In sexually reproducing organisms, offspring inherit a mix of characteristics from two parents.

FIGURE 3.21 Genetic continuity at the species level involves conservation of diversity of traits.

In both cases, the correct number of chromosomes and characteristics are passed on in family lineages. At a *genetic level*, stability arises when chromosomes are replicated accurately to give rise to identical daughter chromosomes.

For continuity *at the species level*, successful desirable traits must be passed on, along with some random errors (chance errors may be introduced by mutation). This allows a species to evolve if an environmental change occurs. Natural selection acts so that individuals in a population that are best suited to the environment survive and reproduce, passing on their genes to their offspring. This mixing of parent genes during sexual reproduction, including some that have arisen due to mutation, increases genetic diversity and helps maintain continuity of the species.

The mechanisms that have evolved to ensure genetic continuity (passing on of genetic traits) and the survival and continuity of species include:

- consistent replication prior to cell division (mitosis and meiosis)
- an orderly distribution of chromosomes when cells divide and when gametes form
- fertilisation methods that ensure that individuals of the same species breed successfully
- methods to ensure embryo survival, such as production of large numbers or protection and nourishment of developing embryos and parental care
- natural selection so that the fittest survive to reproductive age and pass on their genes.

Mechanisms that result in genetic variation in species include:

- mutation – changes in DNA due to mutation may be spontaneous or mutagen-induced
- mixing of parental genes during sexual reproduction (brought about by crossing over and independent assortment during meiosis, and random fertilisation of gametes).

Genetic errors that threaten the continuity of species

You have learned that many variations that arise due to spontaneous changes in DNA are temporary and are immediately corrected by enzymes that bring about DNA repair. The number of DNA repair enzymes present in cells is an indication of how important accurate replication and DNA repair are for survival. In humans, a range of diseases have been linked to the reduced ability of cells to repair DNA during or after replication. Research shows that people who have a reduced ability to repair DNA tend to be more susceptible to some cancers. A decrease in the ability of cells to repair DNA during replication is also thought to be responsible for accelerated ageing and may give rise to neurodegeneration. There is a great deal of current research in this area.

When a mutation is present in a DNA repair gene, the gene may be expressed in an altered form or not expressed at all. For example, it has been found that people who have the disease *Xeroderma pigmentosum* (XP) are unable to repair DNA and so they are more vulnerable to DNA damage from ultraviolet rays and, as a result, to skin cancer. Recent research shows a link between germline mutations in DNA repair genes and lethal forms of prostate cancer. Research in animal embryology shows that if one of the genes for DNA repair (the base excision repair gene) is missing, this results in the death of the embryo. Mutations such as these are termed lethal mutations.

Genetic information can only be stored in a stable form and passed on consistently if DNA repair enzymes continuously scan the DNA for errors in replication and replace incorrect or damaged nucleotides. Natural selection is a mechanism that ensures individuals carrying damaged genes are removed from populations so that the continuity of species is not at risk.

INVESTIGATION 3.3

Literature review of the effect of cell replication processes on the continuity of species

Literacy

Information and communication technology capability

TASKS

A Assess the effect of cell replication processes on the continuity of species.

B Present your findings in the form of a literature review.

C Acknowledge and evaluate your sources.

You will complete secondary-source research to review the effect of *accurate replication* as opposed to *inaccuracies in replication* on the continuity of species.

PART A: RESEARCH

Focus question: In what ways do cell replication processes support or threaten the continuity of species?

1. Draw up a chart in which you list the following in relation to the focus question:
   - What you know: list any facts relevant to the topic that you have learned during this section of the course.
   - What you think you know: list anything based on prior knowledge, or that you think you understand.
   - What you need to find out: outline some research questions (key concepts) and write some key words.
2. Read the articles listed in the weblinks provided and find your own articles, and use these to gather valid and reliable information relevant to your research questions.
3. Interpret and analyse your search results. (You may need to modify your search strategy as you go.)
4. Make a judgement and use your findings to support your concluding judgement in answering the focus question.

Weblink
Threats to genetic continuity

Weblink
What happens when mitosis goes wrong?

Weblink
Cancer-specific defects in DNA repair pathways

Worksheet
Evaluating resources

Evaluate your sources using the CRAAP technique. (See page 10.)

PART B: PRESENT YOUR FINDINGS

5. Write a literature review of approximately 400 words (page 9), with:
   - an *introduction*, where you define the topic
   - a *body*, where you group the literature and your findings according to common themes
   - a *conclusion*, where you explain the link between your research question and the literature you have reviewed, creating an evidence-based argument.

PART C: ACKNOWLEDGE SOURCES

6. Acknowledge and evaluate your sources using an accepted referencing style.

KEY CONCEPTS

- Genetic continuity relies on:
  - consistent replication of genetic information that is passed from a parent cell to daughter cells, resulting in continuity in the traits being passed from parents to offspring
  - the effect of natural selection and evolution on the gene pool as a result of:
    - introduction of variation during sexual reproduction
    - random errors arising by mutation, being replicated and passed on to offspring.
- Random variations that confer an advantage may be selected over those that confer no advantage or are harmful.

CHECK YOUR UNDERSTANDING 3.5

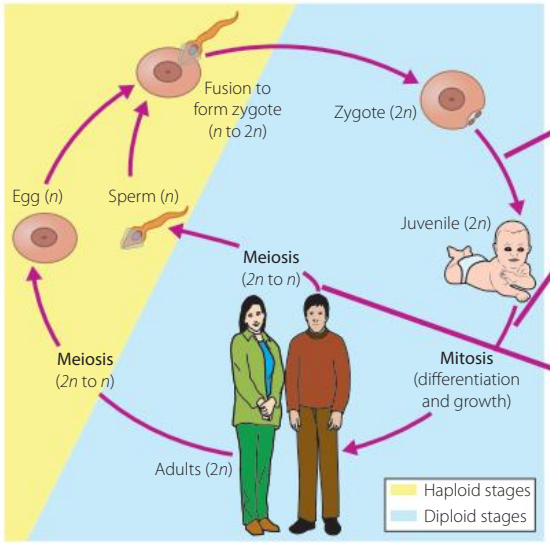
1. Explain the role of DNA replication in:
   - **a** maintaining genetic continuity in a species
   - **b** introducing genetic variation in a species.
2. Explain how natural selection can remove harmful variations from a species.

3 CHAPTER SUMMARY

Cell replication: How important is it for genetic material to be replicated exactly?

PROCESSES INVOLVED IN CELL REPLICATION: MITOSIS AND MEOISIS

Roles in life cycle



Importance of mitosis and meiosis

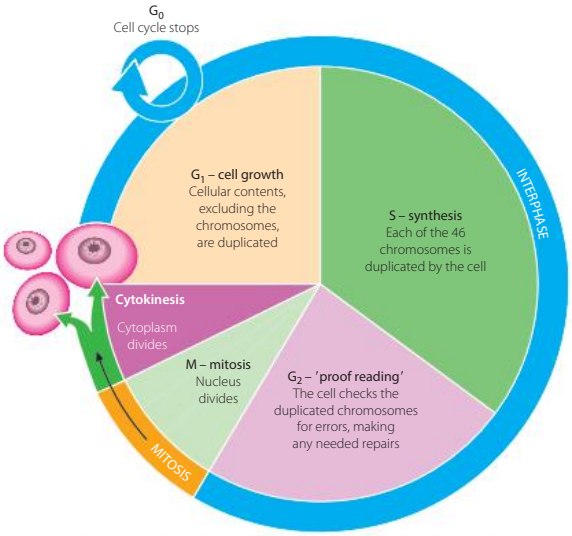
Mitosis

- Embryonic development: zygote ⟶ blastula ⟶ embryo
- Growth of multicellular organisms – meristem (plants); stem cells (animals)
- Tissue maintenance and repair
- Asexual reproduction and genetic stability in populations

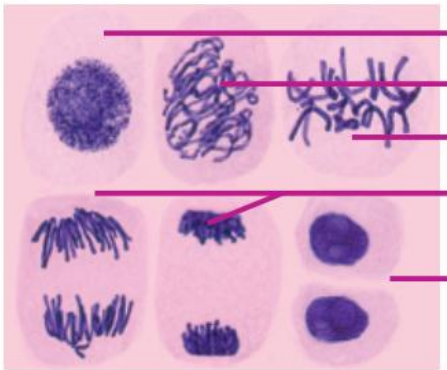
Meiosis

- Production of gametes
- Halving the chromosome number
- Introduction of genetic variation

The cell cycle



Stages of mitosis



Interphase – DNA replicates

Prophase – chromosomes appear and split into chromatids

Metaphase – chromosomes align on equator

Anaphase & telophase – daughter chromosomes segregate and move to poles

Cytokinesis – cytoplasm divides

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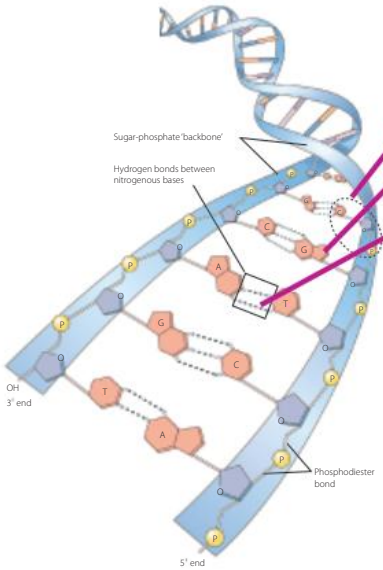
DNA STRUCTURE AND REPLICATION: THE WATSON CRICK DNA MODEL

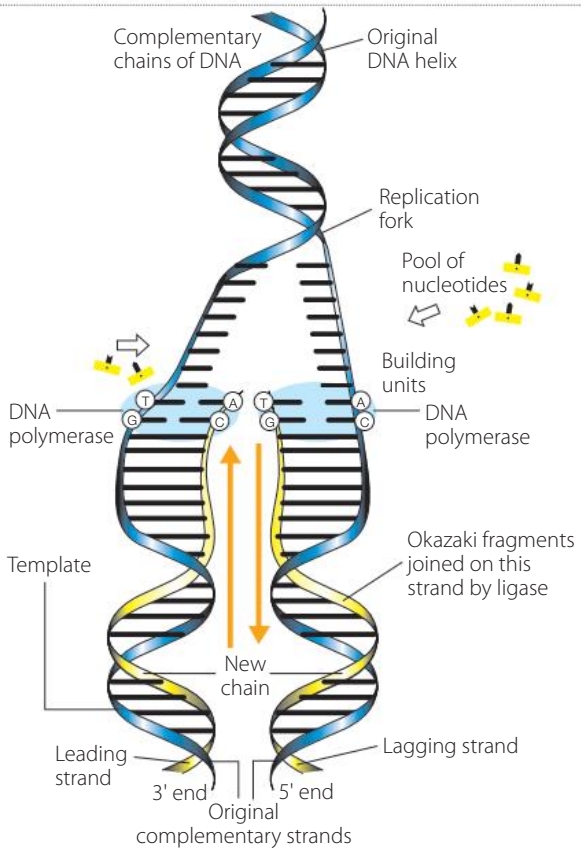
Nucleotide composition, complementary base pairing and bonding



Nucleotide = phosphate – sugar – nitrogenous base

Nitrogenous base pairing:
adenine + thymine; cytosine + guanine

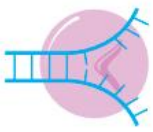
Bonds = weak hydrogen



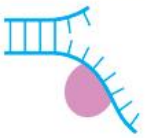
Semi-conservative DNA replication

- DNA unzips and a replication fork can be seen.
- Each strand acts as a template.
- DNA polymerase adds nucleotides from 3′ to 5′ end.
- New molecules consist of one old and one new strand.

Ensuring exact replication (enzyme regulation)

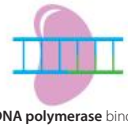


Helicase unwinds parental double helix.



Binding proteins stabilise the strands.

Primase adds a short primer to template strand.

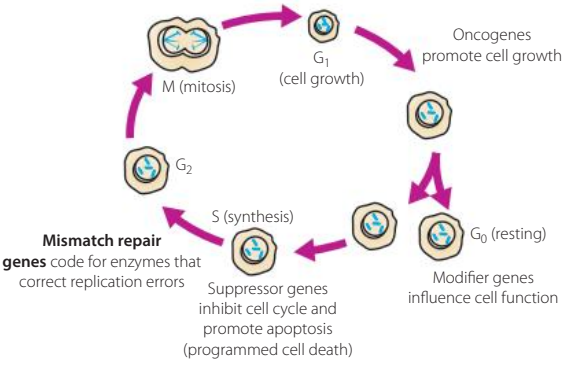


DNA polymerase binds nucleotides to form new strands.

Exonuclease removes RNA primer and inserts correct bases.

Ligase joins Okazaki fragments and seals nicks in sugar-phosphate backbone.

Accurate DNA replication ensures the survival of the species.



3 CHAPTER REVIEW QUESTIONS

Review quiz

1. Name the three types of cell division found in all living organisms and the purpose of each.
2. Identify the components of chromatin.
3. Explain why chromosomes are only visible during cell division.
4. Name the phases of the cell cycle and outline what happens in each phase.
5. Give three reasons why it is important for the cell cycle to be regulated.
6. The graphs in Figure 3.22 show cell volume (the size of a cell) and the amount of DNA in a cell at various stages of the cell cycle.
   a. How many cell cycles (cell divisions) has the cell shown in the graphs completed? Justify your answer.
   b. Explain the change in the amount of DNA during the cell cycle, as shown in the lower graph. Use correct terminology for the phases of the cell cycle and the stages of mitosis.
   c. Draw a sequence of diagrams of a dividing cell (to scale) to show how the cell changes in cell volume during the process of mitosis and use data from the graph to justify your explanation.
   d. Describe the relationship between the cell volume and the amount of DNA in the cell.
   e. The cell volume graph is divided into ten units of time. At what time does cytokinesis occur? (Express this in numbers of time units.) Give a reason for your answer.
7. Distinguish between mitosis and cytokinesis.
8. Explain why it is important for DNA to replicate before cell division.
9. Draw and label a diagram of an RNA nucleotide.
10. State the rule of base pairing of nitrogenous bases in a DNA molecule.

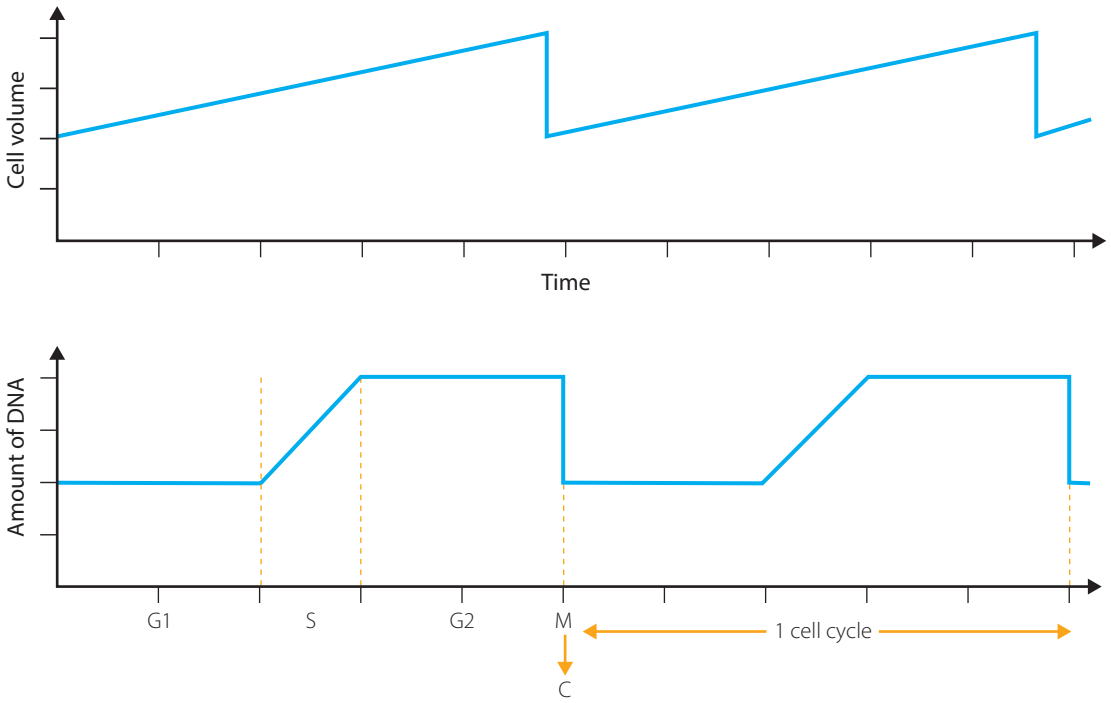


FIGURE 3.22

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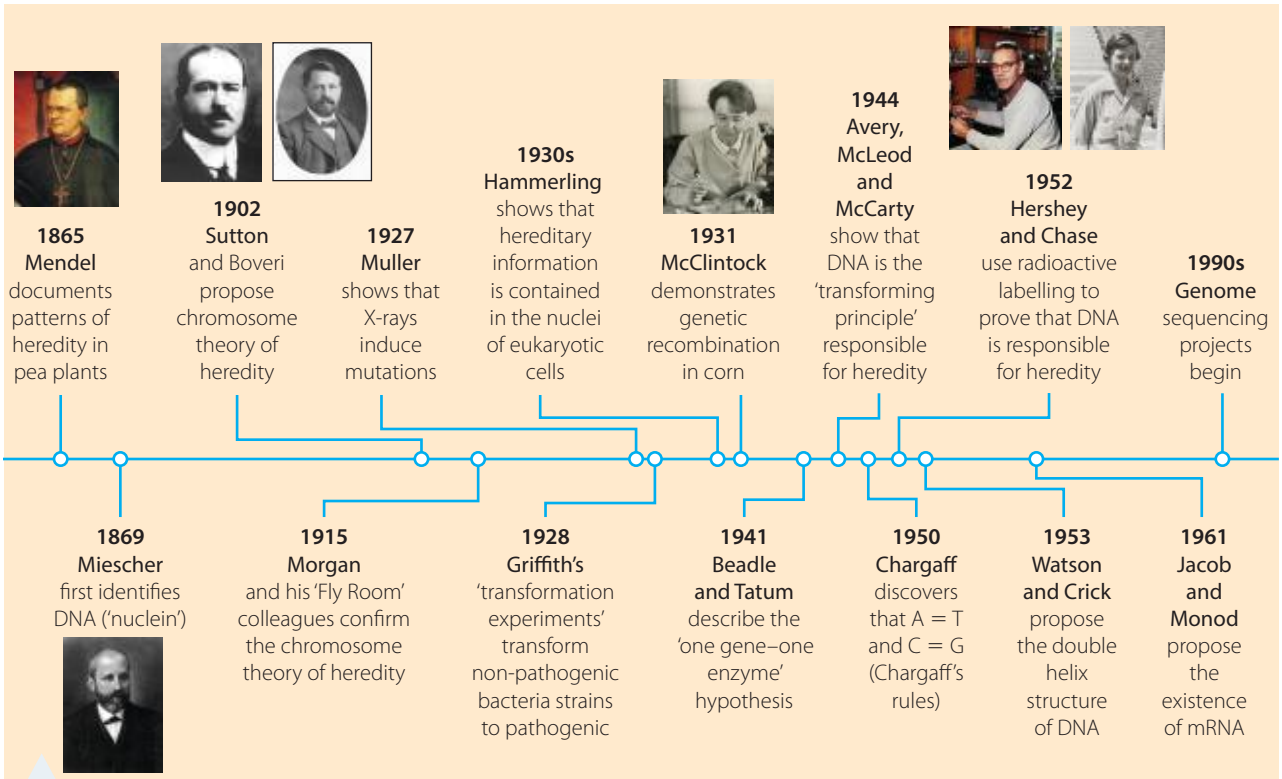
11 Based on the timeline in Figure 3.23, create a table to summarise the contributions made by scientists in the discovery of the hereditary material in cells.



Left to right: Alamy Stock Photo/Pictorial Press Ltd; Walter S. Sutton Collection, University of Kansas Medical Center Archives, Kansas City, KS; Hulton Archive/Getty Images; PF-(bygone1)/Alamy Stock Photo; Bettmann/Getty Images; Getty Images/Hulton Archive/Stringer; Alamy Stock Photo/PF-(bygone1); Getty Images/Bettmann Science Photo Library; Bottom: Alamy Stock Photo/Paul Fearn; The History Collection/Alamy Stock Photo

FIGURE 3.23 Timeline of contributions by scientists to our understanding of genetics and inheritance

12 List two hypotheses that Watson and Crick tested when creating the models listed below, and explain how their findings supported their hypotheses:
- **a** DNA structure
- **b** DNA replication.

13 Explain how nucleotides are added to both strands during DNA replication. Use a diagram to illustrate your answer.

14 Predict the DNA sequence for a complementary strand of DNA made from the following sequence of bases: AATTGGCTGACGAATCAT.

15 Outline the role of four named enzymes in cell replication.

16 Explain why DNA replication is referred to as 'semi-conservative' replication.

17 Explain, giving examples, the importance of exact replication of DNA in cell division and explain the role that enzymes play in 'proof reading' the molecule after replication.

18 Explain the following features of the Watson and Crick model of DNA, supplementing your explanation with simple diagrams where necessary:
- **a** nucleotide composition
- **b** nucleotide pairing
- **c** nucleotide bonding.

19 Assess the effect of the following processes on the continuity of species:
- **a** DNA replication
- **b** action of polymerase enzymes during DNA replication
- **c** separation of daughter chromatids during mitosis
- **d** formation of identical cells in multicellular organisms
- **e** cell replication in unicellular organisms
- **f** replication of cell organelles during G_1 phase.

20 Answer the inquiry question at the start of this chapter: How important is it for genetic material to be replicated exactly? Use information from this chapter to support your answer.

Exam preparation

4 DNA and polypeptide synthesis

INQUIRY QUESTION

Why is polypeptide synthesis important?

Students:

- construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes (ACSBL076) ICT
- model the process of polypeptide synthesis, including: (ACSBL079)
  - transcription and translation
  - assessing the importance of mRNA and tRNA in transcription and translation (ACSBL079)
  - analysing the function and importance of polypeptide synthesis (ACSBL080)
  - assessing how genes and environment affect phenotypic expression (ACSBL081) CCT L
- investigate the structure and function of proteins in living things L

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
- **4.1** Modelling DNA in prokaryotes and eukaryotes
- **4.2** A practical and secondary-source investigation to model polypeptide synthesis
- **4.3** A secondary-source and practical investigation into the effects of environment on gene expression and resulting phenotype
- **4.4** Secondary source investigation to assess how genes and environment affect gene expression
- **4.5** Secondary-source investigation into protein structure and function

Worksheets
- The process of protein synthesis
- Protein synthesis presentation
- Extension: Investigating alcoholism, IQ and height
- Epigenetics: chemical modification of gene expression that may be inherited
- Protein revision

Nelson MindTap

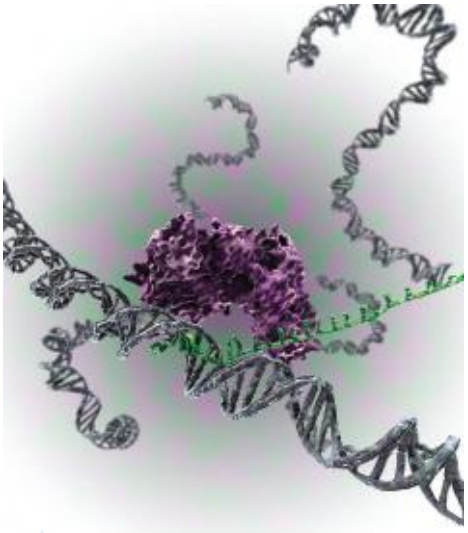
To access these resources, visit **cengage.com.au/nelsonmindtap**

Have you ever wondered how our genes give us visible features? How they play a part in determining characteristics such as height and build, eye colour and skin colour – and what influence, if any, the environment has? To understand the continuity of life at the organism level, we need to explore how genes translate into physical and behavioural features in our bodies and chemical structures such as proteins in our cells.

As you learned in the previous chapter, the continuity of life describes how new organisms arise from living organisms of the same type. The continuity of life is maintained at the cellular level by cell division (mitosis and meiosis) and at the molecular level by DNA. At the whole-organism level, continuity of life depends on physical and chemical features in organisms that result when the instructions in DNA are translated into proteins.

Questions in science drive research. After the structure and mode of replication of DNA were discovered, the next questions asked were:

- Is the genetic code universal?
- Are the structure and function of DNA in prokaryotes the same as in eukaryotes?
- How does the coded sequence of nucleotides in DNA produce proteins?



Gunilla Elam/Science Photo Library

FIGURE 4.1 DNA base sequences are expressed as proteins.

Research into gene functioning continued after the development of the Watson and Crick model and the more scientists learned, the more they wanted to find out. Scientists began investigating **gene regulation** – how and why some genes 'switch on' to produce a product in some cells and not in others. For example, why is it that skin cells make the pigment melanin, but bone cells do not? And why is it that bone cells make bone, but skin cells do not?

In this chapter you will study DNA in prokaryotes and eukaryotes, and how DNA codes for proteins.

4.1 DNA in prokaryotes and eukaryotes

The basic principles of molecular biology and genetics apply to both prokaryotes and eukaryotes, and genetic evidence points towards all present-day cells having evolved from a common ancestor. The genetic code is universal – the same nucleotide base-pairing code is used in all living organisms, both prokaryotes and eukaryotes, to instruct protein synthesis. Both types of cells use a similar mechanism to translate information from DNA into polypeptides and proteins within the cell.

The basic principles learned from experiments performed with one type of cell have been found to apply to other cell types. This has led to a common model system used in the study of molecular biology,

whereby the study of the functioning of genes in simple cells is also applied to more complex organisms. The bacterium *E. coli* is commonly used to study genetics at the molecular level. These bacteria grow easily, divide every 20 minutes when kept at 37°C and model the same type of translation (how mRNA is decoded for the production of proteins) as eukaryotic cells. The metabolism of *E. coli* is very precisely regulated and so it is an ideal example for molecular biologists to use in experiments to research how genes are regulated and expressed. The main differences between bacterial genetics and that of eukaryotes is at the level of **gene expression** (how instructions in DNA are converted into a product such as a protein).

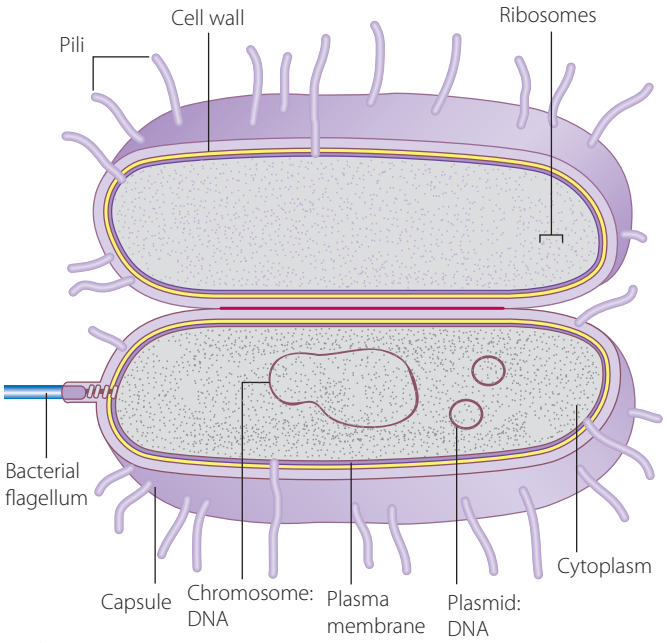


FIGURE 4.2 A prokaryotic cell, with DNA in a chromosome and in plasmids

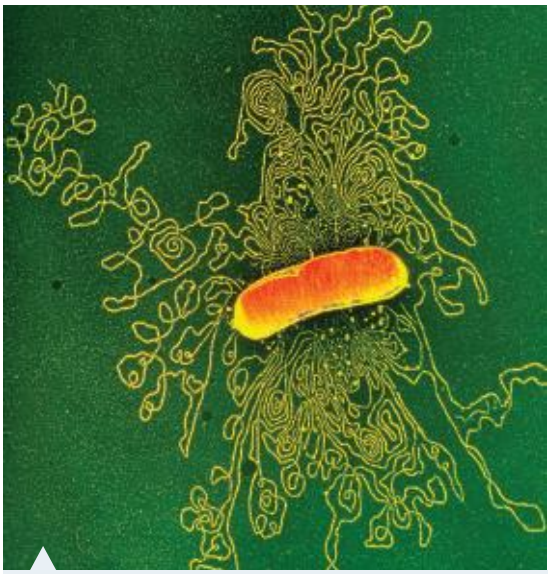
Prokaryotic DNA

The DNA of prokaryotes and eukaryotes is chemically the same, but it differs in how it is structurally arranged into chromosomes and packaged inside the cells. Its location inside these cells is also different, and there are slight differences in the processes of DNA replication and **transcription** (how instructions in DNA are used to make mRNA and initiate protein synthesis).

Location and structure

As you learned in Year 11, prokaryotic cells are 'primitive' cells with a much simpler structure than eukaryotic cells. They contain a single chromosome in the form of a circular strand of DNA (Fig. 4.2). This chromosome has no membrane around it and floats in the cytoplasm, in a dense region known as the **nucleoid**. The DNA codes for proteins that will be made on ribosomes in the surrounding cytoplasm.

The circular, double-stranded prokaryotic DNA is not a helix, but two circles of single-stranded DNA twisted around each other like two pieces of string, each joining at its own ends. The direction and number of twists contributes to the coiling and supercoiling of circular DNA.



Dr. Gopal Murti/Science Photo Library

FIGURE 4.3 Electron micrograph of a ruptured *E. coli* showing a nucleoid – looping strands of DNA around a central protein (scaffold)

Non-chromosomal DNA

Prokaryotic cells may have one or more small rings of non-chromosomal DNA, called **plasmids**, floating separately in the cytoplasm (Fig. 4.2). The genes on these plasmids code for features that are not essential to the survival of the cell, but often provide bacteria with a selective advantage, such as resistance to antibiotics. Plasmids replicate independently of the chromosome.

Packaging of prokaryotic DNA

The circular chromosomal DNA of prokaryotes is about 1300 µm in length. It needs to fit into a cell such as *E. coli*, which is only about 3 µm in length. When *E. coli* DNA is isolated intact, the circular DNA is found to be supercoiled and forms loops around a central protein to form a nucleoid. The dense protein differs from the histone proteins that package DNA in eukaryotic cells. The dense protein is sometimes referred to as the **scaffold** (Fig. 4.3).

Prokaryotes have evolved over millions of years, having been exposed to many and varied selective pressures. Scientists believe

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that, as a result, they have evolved a more refined mechanism for regulating gene expression (an operon system) than complex eukaryotic organisms such as humans.

Eukaryotic DNA

Eukaryotic DNA is located in a membrane-bound nucleus within the cell. Individual DNA molecules are arranged into a number of separate chromosomes, and each chromosome is larger and more complex than the chromosome in a prokaryotic cell. The DNA of a single chromosome in the fruit fly *Drosophila* is 1.2 cm in length. Each human cell contains approximately 2 metres of DNA arranged as a total of 46 chromosomes.

Although eukaryotes have many more chromosomes than prokaryotes, the number of chromosomes is not a measure of how complex the organism is. For example, the Chinese giant salamander has 60 chromosomes, but is less complex than a human (Table 4.1).

TABLE 4.1 Chromosome number in eukaryotes

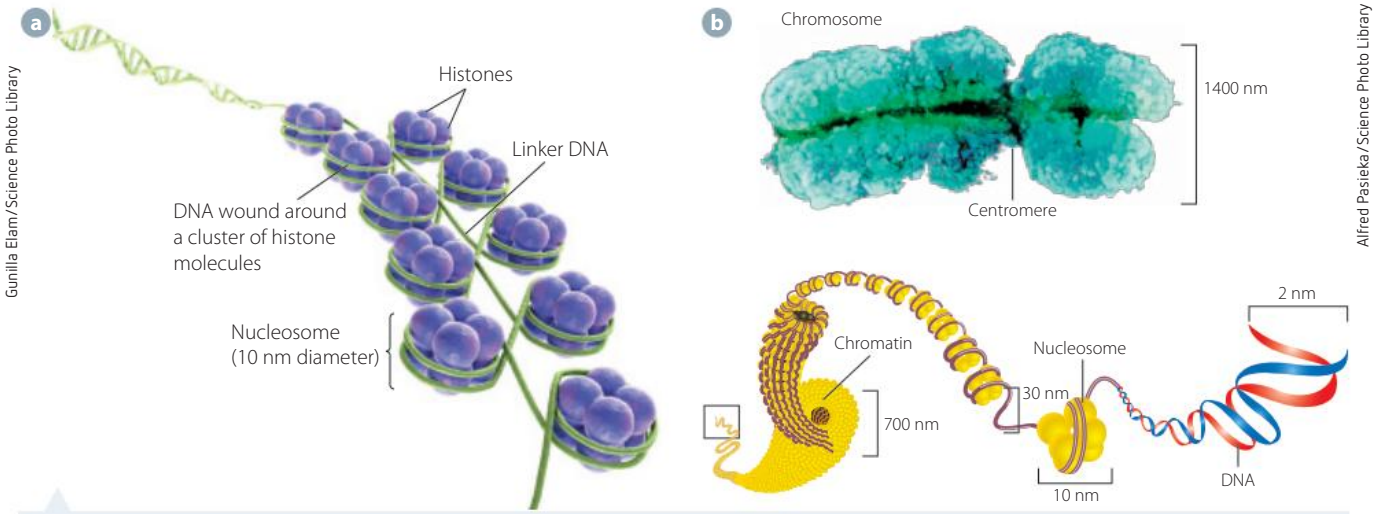
ORGANISM	NUMBER OF CHROMOSOMES (2*n*)
Fruit fly (*Drosophila melanogaster*)	8
Eastern grey kangaroo	16
Earthworm	36
Cat	38
Peanut	40
Human	46
Orangutan	48
Platypus	52
Sheep	54
Chinese giant salamander	60
Horse	64
Black mulberry	308

In most eukaryotic cells there is a large proportion of **non-coding DNA** (DNA that is not used directly to make products such as proteins or RNA) in sequences called **introns**. In humans, only 3% of DNA is **coding DNA** (DNA that contains sequences that code for products such as proteins or RNA). These coding sequences in DNA are called **exons**. The exact function of non-coding DNA is still being researched; it is thought to play a role in the spatial organisation of genes as well as in the control of gene expression. Introns are almost never found in prokaryotes. There are two schools of thought on this – introns may have accumulated during the evolution of eukaryotes, or they may have been lost from prokaryotes as they evolved, simplifying their genome to allow them to divide rapidly.

Packaging of eukaryotic DNA

The DNA of eukaryotes is linear rather than circular and it also winds around proteins (called histones) tightly, but it does not supercoil. It coils in a way that forms nucleosomes – bead-like structures made up of long sequences of DNA wrapped around eight histone protein cores, similar to the way cotton is wrapped around a cotton reel (Fig. 4.4). Unlike nucleiods, which have only one type of protein, there are five main types of histones in eukaryotic cells, and all play a role in packaging DNA. DNA has a series of folding patterns, around different histones. Histones contain a large number of positively charged amino acids, which allow them to bind to the negatively charged phosphates of DNA. As a cell progresses through the cell cycle, the nature of chromatin changes (page 84 and Fig. 5.3, page 152) and these changes are thought to be linked to histone binding.

The effect of histone binding on gene expression is dealt with on page 131 (Fig. 4.21).



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Alfred Pasieka/Science Photo Library

FIGURE 4.4 Eukaryotic DNA: **a** DNA coiled into nucleosomes; **b** top: a chromosome, showing chromatin; bottom: nucleosomes condensed into chromatin

Non-nuclear DNA in eukaryotes

Mitochondria and chloroplasts are organelles in eukaryotic cells that contain their own DNA. This DNA, referred to as **non-nuclear DNA**, is inherited independently of nuclear (chromosomal) DNA. Non-nuclear **mitochondrial DNA (mtDNA)** is found in the respiratory organelles of cells. The discovery of mtDNA has proved extremely useful in studies of evolutionary relatedness.

mtDNA can be used to trace maternal inheritance. During sexual reproduction, the egg and sperm each contribute half the zygote's total DNA. However, sperm cells have very little cytoplasm and the larger egg cell therefore contributes all the cytoplasm (and the organelles within it, including mitochondria). Because mitochondria have their own mtDNA and replicate independently of the nucleus, all mitochondria in a female lineage possess identical mtDNA (unless there is a mutation).

Mitochondrial DNA (mtDNA) is a very small (70 nm in diameter), circular molecule with only 37 genes (Fig. 4.5). Thirteen of these genes make proteins that function in the electron transport chain during chemical respiration in mitochondria. The other 24 genes make RNA molecules (22 make tRNA and 2 make rRNA, which you will learn about later in this chapter).

Each mitochondrion contains about 5–10 circular DNA molecules and each cell has between 100 and 1000 mitochondria. As a result, small samples of tissue yield a large amount of mtDNA. This DNA is easy to sequence because it is so short (16 500 base pairs in humans), compared with nuclear DNA (around 3 billion base pairs in humans).

The rate of mutation of mtDNA is about ten times that of nuclear DNA.

Sequencing of mtDNA is used more often than sequencing of nuclear DNA, because the increased variability of mtDNA makes it very useful for evolutionary studies (such as relatedness testing) as well as for forensic biology (identity testing) and human ancestry studies.

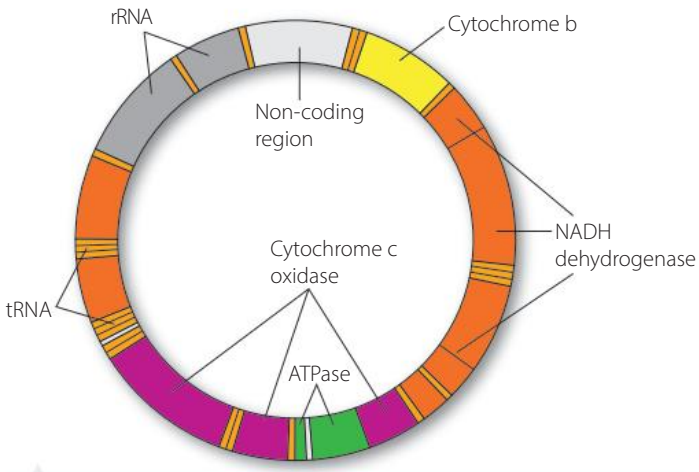

FIGURE 4.5 A model of a mitochondrial DNA molecule, showing some genes (70 nm diameter)

Weblink
Human ancestry testing
Read about mtDNA used to determine human ancestral lines.

Weblink
Forensics and mtDNA
Read about mtDNA used in human identification.

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The use of mtDNA is of advantage, because mitochondria:

- are inherited only from the mother, which allows tracing of a direct genetic line
- do not combine paternal and maternal genes, as nuclear DNA does (mixing genes during gamete formation and fertilisation)
- occur in large numbers in every cell, so they are easy to access and sample
- evolve very quickly because mtDNA does not have repair enzymes, and so mutations arise often during replication.

You may wonder how, if mtDNA can undergo so many mutations, the mitochondria are still able to function. Most changes in mtDNA are in sequences that do not code for proteins and so those mutations are usually not harmful. In some instances, if mutations occur in DNA that does code for respiratory proteins, mitochondrial disease may result. (See the weblinks for more information.)

Mixing of paternal and maternal genes during crossing over in meiosis is dealt with in Chapter 5.

Weblink
Mitochondrial disease
Read about mitochondrial disorders, genetic testing and diagnosis.

KEY CONCEPTS

- Nuclear DNA is present inside the nucleus of each of our cells, and has about 3 billion base pairs and around 20 000 protein-coding genes.
- The mitochondrial genome exists outside the cell nucleus, and has 13 protein-coding genes, 24 genes coding for RNA and about 16 500 base pairs.
- mtDNA has a higher rate of mutation than nuclear DNA, making it easier to identify differences between closely related individuals.
- mtDNA is used to study evolutionary relatedness, construct evolutionary trees, investigate family relatedness and identify people in forensic science.

INVESTIGATION 4.1

Modelling DNA in prokaryotes and eukaryotes

INTRODUCTION

Information and communication technology capability

Scientists have used several kinds of cells and organisms as models, to try to investigate and explain aspects of molecular biology such as DNA structure and replication, and its role in gene expression. In simple cells or organelles that have their own short, single molecule of DNA, the features of DNA are much easier to study. Initially molecular biologists used these simple cells as experimental models to work out how DNA functioned. As technology improved and biologists gained a greater understanding of DNA structure and functioning, they moved on to studying similar processes in more complex cells to find out whether they followed the same pattern.

Table 4.2 outlines the features of some simpler cells and organelles that make them particularly advantageous as experimental models.

TABLE 4.2 Genomes used as models for studies of molecular genetic mechanisms, compared with the human genome

	mtDNA (HUMAN)	PROKARYOTE (*E.coli*)	YEAST (*S. cerevisiae*)	EUKARYOTE (HUMAN)
Genome size (base pairs)	± 16 500	4.6 million	12 million	± 3 billion
Number of genes	37	± 4 300	± 6 000	± 20 000
Types of proteins encoded	13(and 24 RNAs)	4 000	5 000	100 000
Number of chromosomes	1	1	16	46
Significance of genome for study	Maternal inheritance allows study of direct lineages. High rate of substitution mutations makes it easier to distinguish between individuals.	Small size makes analysis easy.	Three times larger than *E.coli*, but much simpler than humans.	Complex and large amounts of non-coding DNA makes it more difficult to study.

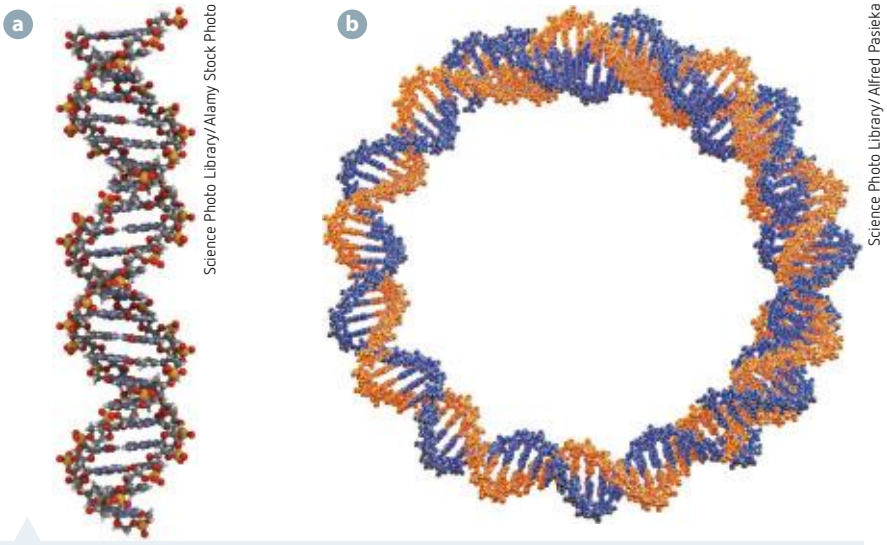

FIGURE 4.6 **a** Linear DNA of eukaryotes and **b** circular DNA of prokaryotes

Weblink
Cells as experimental models for molecular biology
Prokaryotes such as *E. coli*, through invertebrate eukaryotes such as yeast, fruit flies and many others

Weblink
Comparison of prokaryote and eukaryote DNA

Weblink
The complexity of eukaryotic genomes

TASK

1. You are required to use a model to describe, simplify, clarify and/or provide an explanation of the structure of DNA in eukaryotes and prokaryotes. Make sure you consider which features of DNA you intend demonstrating in your model (benefits of the model) and which features you will not be showing (limitations of the model).
2. Write up your investigation as a scientific report, under the headings: Aim, Materials, Risk assessment and safety, Method, Results (the model itself), Discussion (benefits and limitations of the model) and Conclusion.
3. Peer review the models produced by at least two other groups of students. Using what you have learned from these models, the information you have researched and information in this textbook, draw up a table to compare the forms in which DNA exists in eukaryotes and prokaryotes.

   Compare DNA under the following headings:

   - Chromosomal structure
   - Packaging
   - Genetic information stored.

RESOURCES

Use the weblinks as a starting point for your research.

EXTENSION

In your model, include one aspect of the functioning of DNA (such as replication and/or the start of polypeptide synthesis).

KEY CONCEPTS

- Prokaryotic DNA is a circular, double-stranded DNA molecule, supercoiled to form a nucleoid and found in the cytoplasm.
- Eukaryotic DNA is a linear double-stranded helix wound around histones to form nucleosomes.
- Eukaryotic cells also have non-nuclear DNA, such as mtDNA in mitochondria in the cytoplasm. Prokaryotic cells have non-chromosomal DNA in the form of plasmids.

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CHECK YOUR UNDERSTANDING 4.1

1 Explain the meaning of the following terms:
   - **a** gene expression
   - **b** genome
   - **c** nucleosome
   - **d** nucleoid.
2 Explain what is meant by the statement: 'The genetic code is universal'.
3 Give one similarity and two differences between prokaryotic and eukaryotic chromosome structure.
4 Compare the structure and functioning of non-chromosomal DNA in eukaryotes and prokaryotes.
5 Calculate the length of one human chromosome in comparison with the chromosome of a fruit fly.

4.2 Polypeptide synthesis

In the 1950s, Francis Crick proposed that DNA led to the formation of RNA, which in turn led to the synthesis of proteins. Experimental evidence supported his proposal. The 'flow of genetic information' became known as the 'central dogma of molecular biology' (Fig. 4.7). In 1961, Francis Crick and Sydney Brenner provided the missing link to decoding DNA, with their discovery that genes use three-letter 'words' or triplets of bases called **codons** to code instructions for each amino acid in a protein chain.

FIGURE 4.7 The central dogma of molecular biology – a summary of the flow of genetic information resulting in gene expression

Scientists already knew that polypeptides were chains of amino acids and that these polypeptides joined to form proteins. It took about five more years to reveal specifically which triplet coded for which particular amino acid. In 1968, Marshall Nirenburg received a Nobel Prize for his work in cracking the genetic code for protein synthesis, listing the 60 triplets that code for each of the 20 amino acids in proteins (Fig. 4.15, page 125).

A polypeptide is a molecule made up of a chain of many amino acids, joined by peptide bonds (Fig. 4.8). There are about 20 different amino acids that can be linked together in a linear sequence, to form chains of up to 300 amino acids in length.

Assumed knowledge: refer to *Biology in Focus Year 11*, Chapter 3, Section 3.2, Cell requirements: proteins and nucleic acids.

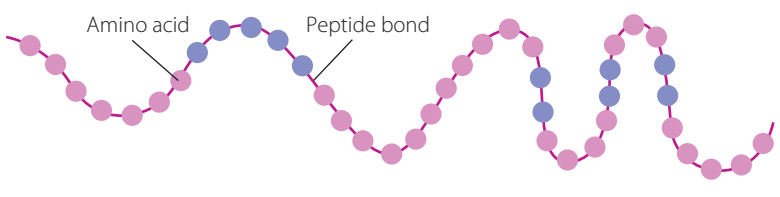

FIGURE 4.8 Amino acids joined together by peptide bonds form the polypeptide chains that make up proteins.

One or more polypeptides twist and join together into a particular three-dimensional shape, forming proteins in cells. The sequence and arrangement of amino acids determines the configuration of the protein (Fig. 4.9). Any change in the amino acid sequence may result in a change in the shape of the protein molecule and this could affect the ability of the protein to carry out its function in the cell.

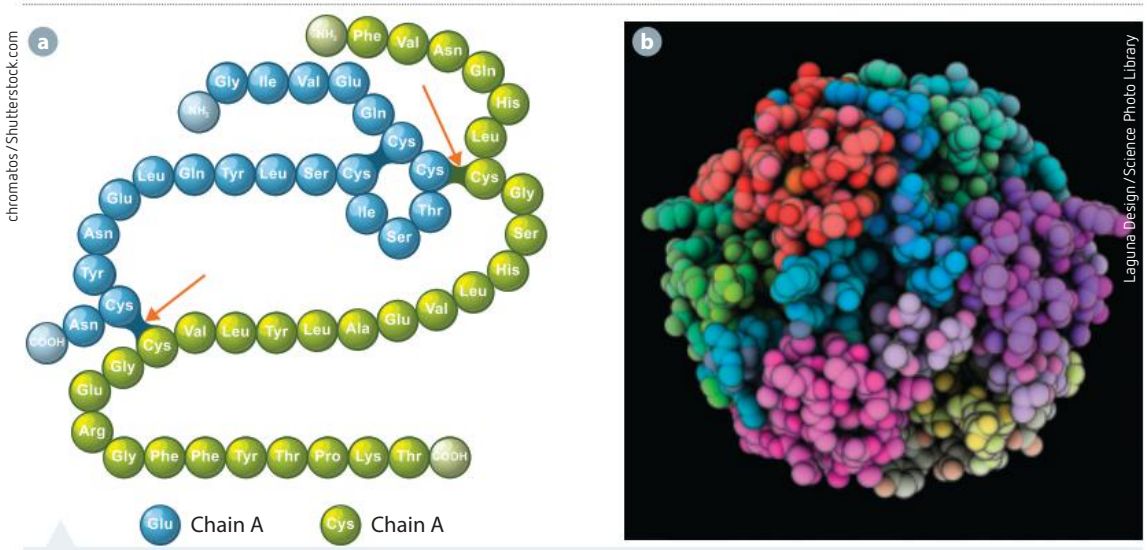

FIGURE 4.9 A protein, the hormone insulin: **a** a simple model showing that insulin is made up of two polypeptide chains joined at the points indicated (see red arrows); **b** a 3D molecular model of the structure of insulin

The 'gene concept' in molecular biology is that genes in a cell contain all the information for the synthesis and functioning of cellular components. When the end product of a gene has been made by the cell, we say the gene has been 'expressed'. In specialised cells in multicellular organisms, only certain genes are expressed in each cell type. Coded instructions for the production of a particular protein (or group of proteins) are said to be 'switched on' in the DNA of that cell. This ensures that each cell develops the necessary structures, in keeping with the type of tissue to which it belongs. For example, in skin tissue, genes for the pigment protein *melanin* and the protein *keratin* are switched on in each cell, ensuring that the cells become skin cells. Different genes are expressed in nerve cells, muscle cells and bone cells (Fig. 4.10).

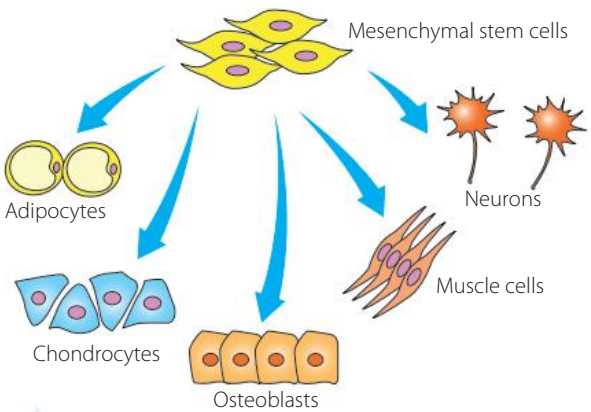

FIGURE 4.10 Cells differentiate and specialise as a result of gene expression.

The process of polypeptide synthesis

DNA never leaves the nucleus – the molecules are too large to pass through the pores in the nuclear membrane. This is just as well, as DNA molecules hold the original copy of all instructions for future generations of cells. In order for a cell to make the particular group of proteins it needs, not all the DNA is needed; only the relevant instructions for proteins required in that cell are accessed in the DNA nucleotide sequence. Because the DNA remains in the nucleus, an intermediate molecule called **messenger RNA (mRNA)** is created and this carries a *transcribed* copy of the relevant instructions from the nucleus to the ribosomes in the cytoplasm. The ribosomes are the cell 'machinery' that subsequently *translates* the message carried by the mRNA into a cell product such as protein (Fig. 4.11).

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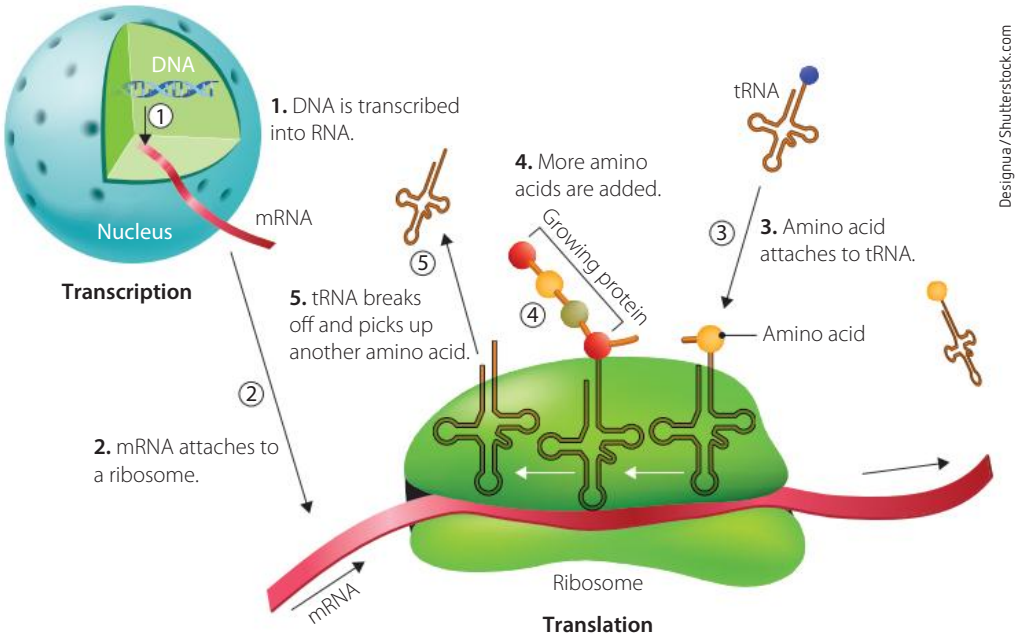

FIGURE 4.11 Gene expression involves the information in DNA being decoded during transcription into RNA (1) and subsequently translated into a product such as a protein (2–4).

Nucleic acids involved in polypeptide synthesis: DNA, mRNA and tRNA

Two types of nucleic acids are essential in the process of polypeptide synthesis: DNA and RNA. There are three types of RNA, each with a specific role to play.

DNA

DNA consists of long chains of nucleotides wound into a double helix. The sequence of nucleotide bases determines the meaning of the message – this is because it codes for the sequence of RNA nucleotides and, ultimately, the sequence of amino acids that form the polypeptide chain.

RNA

Like DNA, RNA is a nucleic acid made up of a chain of nucleotides, but it differs from DNA in the following ways:

- Most RNA is single-stranded.
- The sugar in RNA is ribose sugar (not deoxyribose sugar as in DNA).
- RNA has the nitrogenous base uracil (U) instead of thymine (T).

There are three types of RNA: messenger RNA (mRNA), transfer RNA (tRNA) and ribosomal RNA (rRNA).

- **messenger RNA (mRNA)** is single-stranded and is not twisted into a helix (Fig. 4.12a). mRNA molecules are a few thousand bases long, much shorter than DNA. They are found in both the nucleus and the cytoplasm. mRNA functions as an intermediate molecule, carrying information from DNA in the nucleus to the ribosomes in the cytoplasm.
- **transfer RNA (tRNA)** molecules occur in the cytoplasm. Each molecule is 75 nucleotides long and twisted into the shape of a clover leaf (Fig. 4.12b). At one end of the tRNA are three unpaired bases, called an anticodon, which attach the tRNA to its complementary bases (codon) on the mRNA strand. The other end of the tRNA is able to bind with an amino acid temporarily. Each tRNA molecule will only attach to one particular amino acid. The specific sequence of three bases at the anticodon end determines which amino acid will be carried by that tRNA.
- **ribosomal RNA (rRNA)** forms a structural part of ribosomes (Fig. 4.12c) and is made in the nucleolus of the cell.

Using a computer analogy, you could say that DNA is the operating system, mRNA is the software, tRNA is the machinery (such as a 3D printer) and protein is the product created.

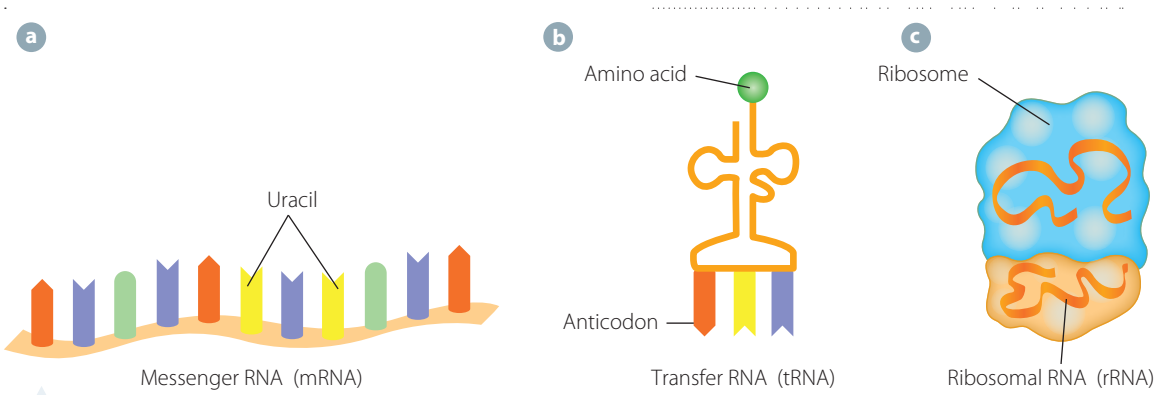

FIGURE 4.12 Three types of RNA: **a** mRNA, **b** tRNA and **c** rRNA (combined with protein to make a ribosome)

Transcription

Transcription occurs when an enzyme, **RNA polymerase**, binds to a section of DNA and begins building a chain of RNA nucleotides to form a complementary strand of RNA. Figure 4.13 shows the details of this process. (The number of each step in the description below matches the sequence of numbered steps in Figure 4.13.)

1 RNA polymerase binds to a part of the DNA called the **promoter** and the DNA 'unzips' – that is, the DNA unspirals, hydrogen bonds between the two strands break, and the strands separate over a short length. This happens only in that part of the DNA that contains the gene to be used. Only one strand of DNA contains the genetic information to make a protein; rather confusingly, it is called the **non-coding strand** or **template strand**; the other strand is called **the coding strand** (it has the same code as the mRNA being made) or **non-template** strand.

2 **Transcription** of the gene is controlled by the enzyme RNA polymerase. The non-coding strand of the DNA acts as a template and RNA nucleotides are assembled, forming a complementary single-stranded mRNA molecule (that is, DNA is *transcribed* into mRNA). The sequence of nucleotide bases on the mRNA molecule is the same as the DNA coding strand or non-template strand, except that it has U instead of T. (In eukaryotes, 'editing' or splicing of pre-mRNA may take place at this point. This is dealt with in more detail on page 124.)

3 The mRNA moves out of the nucleus and into the cytoplasm, where it encounters some of the millions of ribosomes in the cell. Usually one mRNA molecule is read by a large number of ribosomes, so multiple chains of the same polypeptide product are produced from one mRNA template molecule.

Translation

4 Translation occurs when the **ribosomes** move along the mRNA molecule and, as they do so, they attach tRNA molecules to mRNA by temporarily pairing the bases of the tRNA **anticodons** with their complementary triplets of bases (**codons**) on the mRNA.

5 The amino acids from the tail end of each tRNA are linked to one another by an enzyme to form a polypeptide chain. Each amino acid is then spliced off its tRNA carrier.

6 The tRNAs move away from the mRNA, leaving the growing chain of amino acids, and move back into the cytoplasm where they can pick up another amino acid and be reused.

Weblink
Transcription animation
Work through the interactive animation and make notes as you go, to create a summary of the steps involved.

Weblink
Translation (basic)
Work though the interactive animation and draw a flow chart of the steps involved.

Weblink
Translation (advanced)
Play the interactive animation and make a summary of the steps involved.

Weblink
Translation
Watch the video of translation in real time.

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7 The polypeptide chain is then processed in the cell. It may be joined to one or more other polypeptides and be further processed and folded into the correct shape for its functioning, forming the final protein end product.

8 The mRNA is broken down into its individual nucleotides, which can be reused.

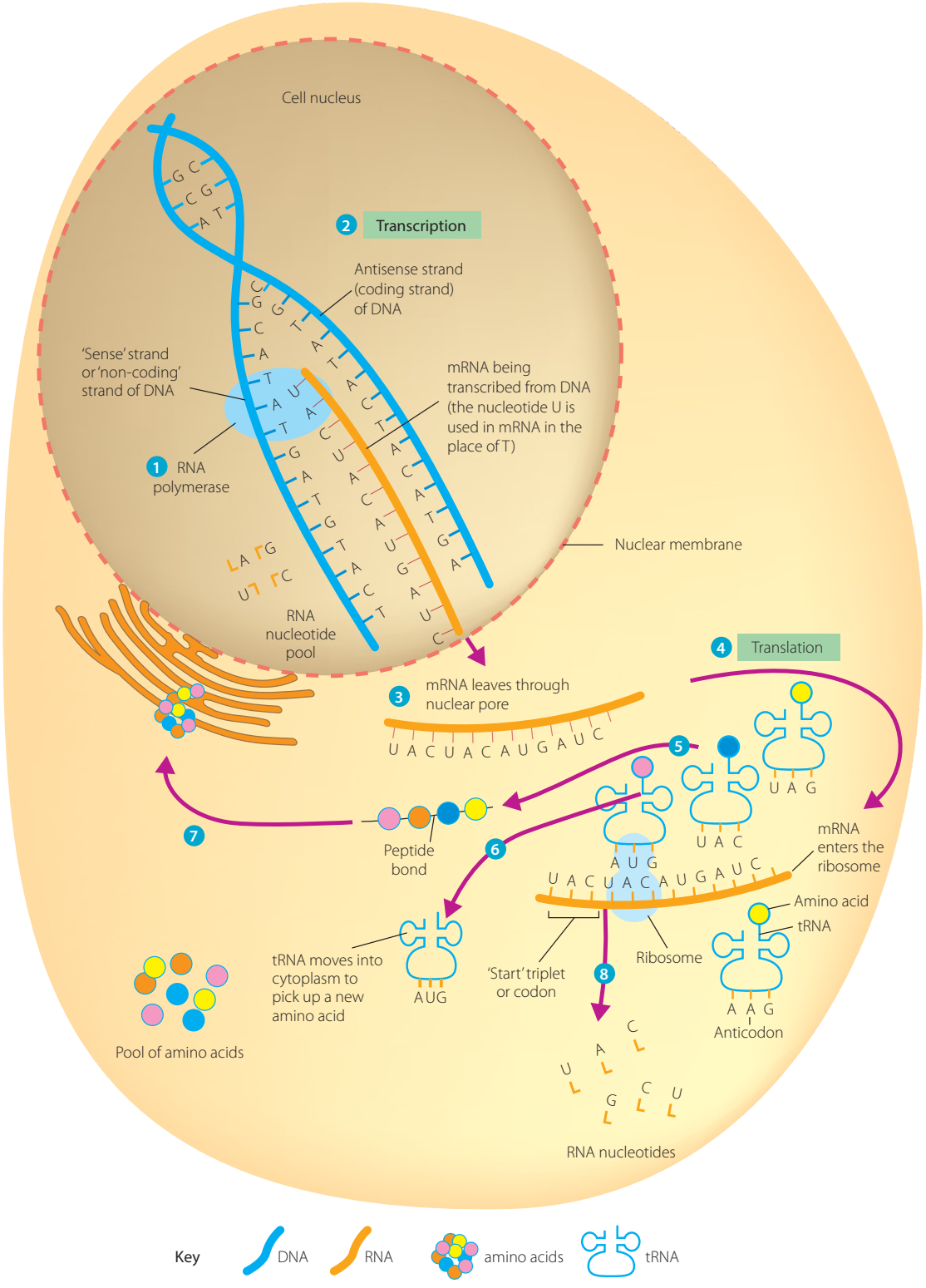


FIGURE 4.13 The biochemical process of polypeptide synthesis in a cell

TABLE 4.3 Types of nucleic acids in cells and their role in polypeptide synthesis

DEOXYRIBONUCLEIC ACID (DNA)	Macromolecule made of a double strand of nucleotides held together by weak hydrogen bonds between base pairs; the ladder-like structure is twisted into a double helix and may be circular (prokaryotes) or linear (eukaryotes); contains the sugar deoxyribose and the bases adenine, thymine, cytosine and guanine
NUCLEAR DNA (nDNA)	DNA arranged as chromosomes in the nucleus of a eukaryotic cell
MITOCHONDRIAL DNA (mtDNA)	Small molecules of DNA found in mitochondria; also known as extra-chromosomal DNA or non-nuclear DNA
RIBONUCLEIC ACID (RNA)	Single-stranded molecule of nucleotides containing the sugar ribose and the bases adenine, uracil, cytosine and guanine
MESSENGER RNA (mRNA)	A single-stranded long RNA molecule containing the sugar ribose and the bases adenine, uracil, cytosine and guanine; it is transcribed from a DNA template (non-coding strand); carries codons (base triplets) that instruct amino acid assembly by ribosomes
TRANSFER RNA (tRNA)	A small RNA molecule folded into a clover shape; carries an anticodon of three bases at one end and a specific amino acid at the other end; works with the ribosome to transfer the correct amino acid for inclusion in sequence to form a polypeptide
RIBOSOMAL RNA (rRNA)	The RNA component of a ribosome which, together with protein, forms the ribosome subunits needed to translate mRNA into a polypeptide chain

KEY CONCEPTS

- Transcription occurs when the double helix DNA unzips and a single strand of mRNA is made, using part of the non-coding strand of a DNA molecule as a template.
- Translation occurs when mRNA is 'read' by ribosomes and translated into a polypeptide, with the help of tRNA.

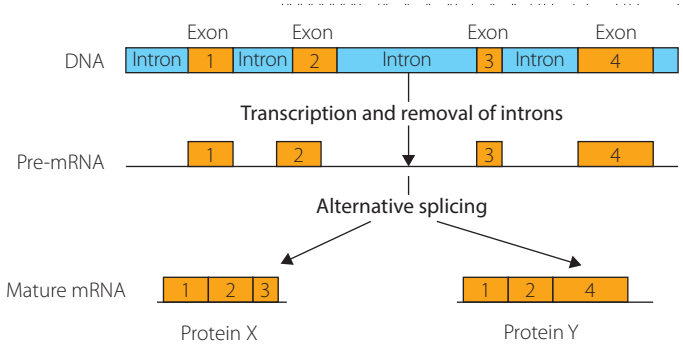
RNA processing

In eukaryotic cells, mRNA that is transcribed from DNA is termed **pre-mRNA**, as further editing of this RNA takes place in the nucleus before it acts as a template for translation into a polypeptide. Pre-mRNA contains coding sequences of nucleotides, called exons, which will be translated into amino acid chains (remember, *ex*ons are *ex*pressed as proteins). What scientists did not realise at first was that in between these exons are sequences of nucleotides called introns, which do not code for amino acid assembly.

Directly after transcription, mRNA is edited (or spliced) and introns are removed (Fig. 4.14) by a complex molecule called a spliceosome. The result of this splicing is the formation of a **mature mRNA** molecule, which then moves from the nucleus to the cytosol for translation by ribosomes. The instructions for splicing the mRNA are found within the introns – they code for their own removal.

Weblink
RNA splicing of introns

FIGURE 4.14 mRNA splicing and gene regulation: following gene transcription, mRNA alternative splicing takes place, whereby non-coding intervening intron sequences are removed by a 'spliceosome' to form mature mRNA.



Splicing is the second step in gene regulation and serves an important purpose in complex organisms. A strand of mRNA produced from one gene is not always spliced in the same way. Alternative ways of splicing mRNA give rise to different versions of the same protein. For example, in humans, immunoglobulins (antibodies) are produced in response to a particular pathogen, such as a bacteria or a virus (pages 398 and 402–403). Within a short space of time, the body produces different forms of antibodies or immunoglobulins specific to the invader. This is done by alternative splicing of mRNA, producing proteins that have a similar structure and/or function but are not identical. These are termed **isoproteins**. Many complex organisms such as vertebrates have up to five times as many proteins in their

Weblink
Alternative splicing
Read about how alterative splicing introduces protein diversity.

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bodies as invertebrates (such as worms and beetles), but only twice as many genes. Alternative splicing provides a mechanism to explain this.

Eukaryotes have a large amount of non-coding DNA, originally termed 'junk DNA' (by Japanese American biologist Susumo Ohno in 1972), as scientists did not know its purpose. For many years, biologists ignored this DNA, but recent studies of non-coding DNA using advanced technology show that its removal disrupts development of individuals in the embryonic state. Further research is showing that non-coding DNA affects the shape of chromatin and how tightly DNA is wound around histones. When some parts of this non-coding DNA are expressed, chromatin is less tightly packed, leading to the likelihood that non-coding DNA determines whether or not certain genes are read. Large portions of non-coding DNA may therefore be involved in regulating which genes are expressed in particular cells, resulting in the differentiation of cells into particular tissue types. The term 'junk' DNA today is reserved only for that part of non-coding DNA that is not involved in gene regulation, DNA for which no known function has been found. With the completion of the Human Genome Project, a large team of researchers has now joined a project called ENCODE to work through the huge amounts of non-coding DNA to find out its function.

As mentioned previously, the universal genetic code of nucleotides in DNA and its transcribed sequence in mRNA determines the amino acid sequence in proteins that are synthesised. Figure 4.15 outlines the nucleotide codons in mRNA that correspond with the 20 amino acids made during translation on the ribosomes. The codon AUG is the start codon, signifying where translation should be initiated. There are three stop codons: UAA, UAG and UGA. The mRNA codon table (Fig. 4.15) can be used to work out the sequence of amino acids that will be created in a polypeptide, based on a DNA and mRNA sequence. Note that there is more than one codon that codes for each amino acid, creating some flexibility for errors in the genetic code.

Non-coding and 'junk' DNA are dealt with in more detail in Chapter 7 on pages 243–4.

Weblink
Is 'junk DNA' our genome's equivalent of a high-level operating system?
Does 'junk DNA' play a role in embryo development? Read about the ongoing debate.

Amino acid key

Ala	=	alanine
Arg	=	arginine
Asn	=	asparagine
Asp	=	aspartic acid
Cys	=	cysteine
Gln	=	glutamine
Glu	=	glutamic acid
Gly	=	glycine
His	=	histidine
Ile	=	isoleucine
Leu	=	leucine
Lys	=	lysine
Met	=	methionine
Phe	=	phenylalanine
Pro	=	proline
Ser	=	serine
Thr	=	threonine
Trp	=	tryptophan
Tyr	=	tyrosine
Val	=	valine

Second base

First base	U	C	A	G	Third base
U	UUU, UUC: Phe UUA, UUG: Leu	UCU, UCC, UCA, UCG: Ser	UAU, UAC: Tyr UAA: Stop UAG: Stop	UGU, UGC: Cys UGA: Stop UGG: Trp	U C A G
C	CUU, CUC, CUA, CUG: Leu	CCU, CCC, CCA, CCG: Pro	CAU, CAC: His CAA, CAG: Gln	CGU, CGC, CGA, CGG: Arg	U C A G
A	AUU, AUC, AUA: Ile AUG: Met/Start	ACU, ACC, ACA, ACG: Thr	AAU, AAC: Asn AAA, AAG: Lys	AGU, AGC: Ser AGA, AGG: Arg	U C A G
G	GUU, GUC, GUA, GUG: Val	GCU, GCC, GCA, GCG: Ala	GAU, GAC: Asp GAA, GAG: Glu	GGU, GGC, GGA, GGG: Gly	U C A G

Use the bases along the sides and top of the table to find the base sequence you are looking for. The cell where the three converge gives the abbreviation of the amino acid that is coded.
Note: Each amino acid may be coded by more than one codon.

FIGURE 4.15 mRNA codon table

INVESTIGATION 4.2

A practical and secondary-source investigation to model polypeptide synthesis

In this investigation, you will gather and process information from several sources to gain an understanding of the process of protein synthesis, and develop your own simple model to explain the sequence of steps involved in the process.

A model of protein synthesis can be developed in a number of ways:

1. class activity: video and group model creation
2. role play: students role play the processes of transcription and translation (also find YouTube video clips on protein synthesis and other relevant websites)
3. assessment task: model and oral presentation
4. computer-generated model.

Weblink
Protein synthesis

PART A: SECONDARY-SOURCE INFORMATION/STIMULUS MATERIAL

Watch the video *Protein synthesis* or another suitable video showing an animated version of protein synthesis. (See the weblink.)

PART B: PRODUCING A MODEL

Working in groups of up to four, use the materials provided to build a working model to demonstrate polypeptide synthesis.

1. Decide as a group:
   - a what materials you will use to depict the structures involved in the process
   - b how you will make the necessary structures out of the materials provided
   - c how the parts will be assembled on the butcher's paper to create your working model
   - d who will make which structures
   - e any colour-coding that may make the model easier to understand
   - f the minimum number of nucleotide sequences you need to use so the resulting polypeptide chain that your model makes contains at least four amino acids.
2. Remember to include in your model a representation of:
   - a both the *nucleus* (or part of a nucleus) where transcription occurs, and the *cytoplasm* where translation occurs
   - b the four different nucleotide bases present in your DNA and RNA strands
   - c ribsosomes, tRNA and amino acids.
3. Labels, arrows and/or numbers are useful to indicate the sequence of events (Fig. 4.16).

Literacy

FIGURE 4.16 Creating a model of protein synthesis

PART C: PRESENTING THE MODEL

Use the model to give a short oral explanation (as a group) of the process of protein synthesis to your classmates. Be aware of the limitations of your model – these will be assessed by the class at the end of each presentation.

You may be asked to explain any of the following concepts, using your model.

1. Outline how a change in DNA sequence can result in changes in proteins produced and therefore changes in cellular activity.
2. Explain how mutations in DNA may lead to the production of a non-functional protein.
3. Explain why the 'one gene – one protein' hypothesis was altered to the 'one gene – one polypeptide' hypothesis.

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PART D: REPORT

Write a report on the model that you and your group presented.

1. Outline the purpose of developing this model.
2. List the materials.
3. Justify the validity of your model.
4. Discuss any limitations of your model.
5. Acknowledge all secondary sources, using an accepted referencing style.

Worksheet
The process of protein synthesis

PART E: DO IT YOURSELF

View the presentation and complete the worksheet, *The process of protein synthesis*, using the mRNA codon table in Figure 4.15 to translate a protein from a given DNA sequence. (See the link provided.)

Worksheet
Protein synthesis presentation

Function and importance of polypeptide synthesis: genes to proteins

A **gene** is considered the smallest unit of heredity. Chemically, each gene is a portion of DNA with a specific sequence of bases that encodes for a particular trait that can be passed from parent to offspring. A **locus** is the position of a gene on a chromosome. The coded information within genes determines how living things look, behave and function – that is, it influences particular characteristics (phenotypes). A chromosome can therefore be described as a linear sequence of genes. The total amount of genetic material that an organism has in each of its cells is called its **genome**.

Specific staining techniques are used to show banding patterns on chromosomes (Fig. 4.17) and these bands correspond on homologous pairs of chromosomes. The banding patterns can also be used to identify the positions of particular genes on chromosomes (Fig. 4.18). With modern technology, particular genes can be marked with fluorescent tags that show up on the chromosome, assisting gene mapping. Specific genes can therefore be associated with a particular physical feature or **trait**. **Alleles** are different forms of the same gene (Fig. 4.18).

Weblink
How much DNA codes for protein?
List the following structures in order of size, from smallest to largest: chromosome, gene, DNA, nucleotide, base, genome.

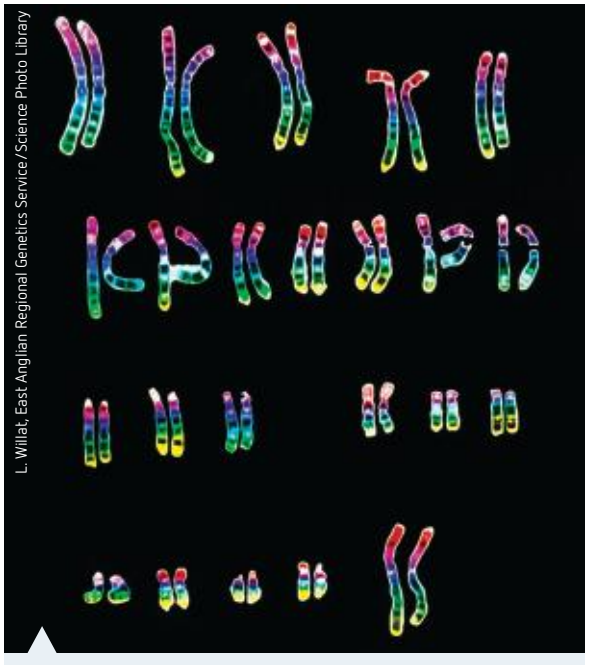

FIGURE 4.17 Human karyotype with 46 chromosomes, showing the banded patterns of gene loci

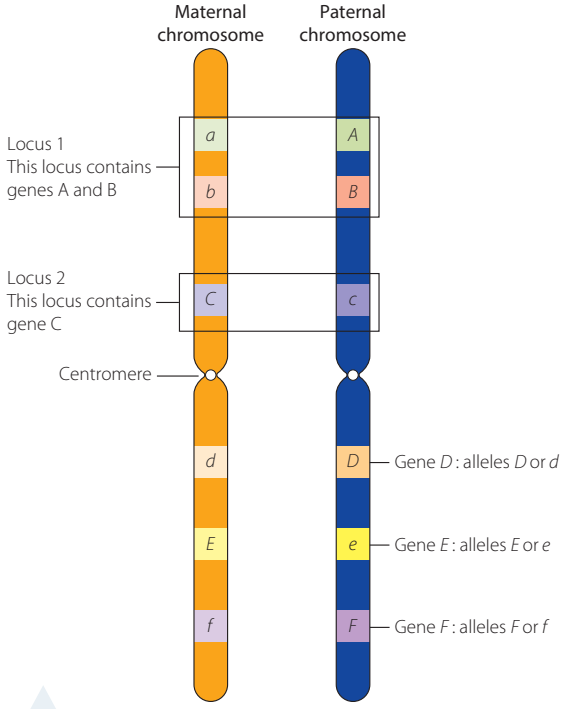

FIGURE 4.18 Genes located on homologous chromosomes

Changing definition of a gene

The definition of a gene has changed as biologists come to understand more about how genes function. At first, a gene was defined as *a sequence of nucleotides that codes for one* **protein**. Advances in understanding of the biochemical functioning of cells have led to the definition changing to *a sequence of nucleotides that codes for one* **polypeptide chain**. More recent research has shown that some genes code for rRNA and tRNA, which are not proteins at all, and that in other instances one gene may code for more than one polypeptide sequence (due to the splicing and rearrangement of blocks of mRNA before translation). Therefore the definition of a gene may need to change to a more functional concept: *a sequence of nucleotides that codes for any molecular cell product.*

KEY CONCEPTS

- A gene is made up of linear sequences of nucleotides that code for a cell product, such as a polypeptide chain.
- In eukaryotes, mRNA may be processed after transcription (introns are spliced out) before it passes into the cytoplasm and binds with ribosomes to direct the formation of a polypeptide.
- mRNA is translated, with the help of tRNA on ribosomes, into polypeptide chains. A table of mRNA codons has been created from researched evidence, showing which triplets of bases code for which amino acid.
- The process of polypeptide synthesis may be modelled, but all models have limitations.
- Homologous chromosomes carry genes for the same traits in the same position (locus), but these genes may have alternative forms (alleles).
- Specific genes are associated with specific traits and are expressed in specialised cells.

CHECK YOUR UNDERSTANDING 4.2

1. Compare DNA and mRNA in terms of structure and function.
2. Distinguish between the three types of RNA, state where each is found and outline the role of each in polypeptide synthesis.
3. The genetic code is sometimes described as a triplet code. Explain what this means.
4. Draw a diagram to show the meaning of the following terms: chromosome, gene, allele, locus.
5. Distinguish between the terms gene, genome and trait.
6. Draw a flow chart to model the process of polypeptide synthesis. Identify two benefits and one limitation of your flow chart model.

4.3 How genes and the environment affect phenotypic expression

The *genotype* is the set of genes in an organism's DNA that is responsible for a particular trait. Genotype can be determined by biological testing. The *phenotype* is the physical expression, or characteristics, of that trait and can be determined by observation.

Is the ability to play sport inherited? Why does tightrope walking tend to run in families? Is shyness genetically determined or does the environment in which we grow up influence this? What about intelligence? Why do identical twins who are separated at birth and later reunited often show unexpected similarities, such as having the same hobbies, liking the same foods and even choosing the same career? If they are genetically identical, why do these twins have differences, such as different susceptibility to certain diseases?

The phenotype of an organism is often defined as its physical appearance, but today we know that phenotype includes the sum of all gene products. The term **phenotype** in this context includes the structure, behaviour and physiology of an organism.

We also know that the genotype (genetic blueprint) of every cell is the same, yet each cell becomes a specific type of cell in a particular tissue. This specialisation of cells is brought about by controlling the expression of genes within the cells. Gene expression is the translation of genes into their protein end

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products. These products determine the physical and chemical features typical of each cell type and the overall phenotype of an organism.

Some variations in organisms are genetically determined ('nature'), whereas others are influenced by the environment ('nurture'). Many variations arise as a result of an interaction between the two – the environment can influence how genes are expressed. The term 'nurture' here refers to a wider influence of the environment than just the nurturing one of the home; it includes all environmental influences.

Gene expression has been studied for over half a century, with biologists intent on finding out how the gene sequence shows up as the phenotype. For example, how does DNA determine what blood group a human belongs to, what colour fur an animal has and how tall a plant grows?

FIGURE 4.19 Identical twins: any differences are due to effects of the environment.

Studies of both plants and animals show that, although genes may be direct determinants of phenotype, gene expression can be enhanced or masked by factors in the environment. In twin studies, the phenotypes of identical **twins** (with identical genotypes) have been analysed. The reasoning behind this is that any phenotypic differences between identical twins must reflect the influence of the environment. Studies of twins separated at birth have formed a major part of these studies, because the effect of 'nature' (genotype) and 'nurture' (environment) can then be explored independently.

More recent research involves biologists investigating how the environment influences gene expression at a molecular level, leading to a field of study called **epigenetics**. Some results suggest that the environment may chemically modify DNA in individuals and in this way affect gene expression. This chemical modification is not a change in the sequence of bases in the genome (as in mutations), but instead seems to involve chemical markers or tags being added to DNA.

Identical twins have identical genes, but may not have identical gene expression, a difference that may be influenced by epigenetics.

Gene expression and phenotype

Genes that are expressed dictate the types of proteins in cells and, as a result, the overall phenotype of organisms. A great deal of current research involves using stem cells to try to find out how cells 'know' what kind of cell to become. **Stem cells** are unspecialised cells that are capable of dividing and becoming specialised tissue. Embryonic stem cells are capable of dividing and giving rise to any type of tissue within an organism, and so they are termed **pluripotent** (*pluri* = many; *potent* = potential). Stem cells also occur in some adult tissue (adult stem cells) but these are not pluripotent, as they are only able to give rise to cells of one tissue type. Stem cells undergo asymmetric division – they give rise to two daughter cells, one of which will continue to divide and another that will follow the path of differentiation. Studies have shown that when stem cells differentiate, special proteins called **transcription factors** appear to control which genes in the cells are transcribed. These transcription factors therefore determine the developmental pathway of a cell and the type of tissue it will become.

It is interesting to note that, because each step in the process of gene expression is regulated by proteins, genes must produce the proteins that regulate their own expression. The accurate synthesis of proteins according to DNA instructions is therefore of ultimate importance in assembling amino acids in the correct order in each polypeptide, as this gives rise to the three-dimensional structure of each protein – essential for the correct functioning of cells and to produce an overall phenotype in organisms that is free of disorders.

Worksheet
Extension: Investigating alcoholism, IQ and height

Weblink
The epigenetics of identical twins
Describe the model that was used to represent epigenetic modification of chromosomes.

Worksheet
Epigenetics: chemical modification of gene expression that may be inherited

Environmental effects on gene expression and phenotype

The coat colour of Siamese cats is determined by a colour mutation (Fig. 4.20a, b). Cats that have the allele *C* have uniform pigmentation of their fur over all parts of their bodies. However, cats that are homozygous recessive for the mutant allele *c* have dark pigmentation at the extremities of their bodies – the tips of their ears, tail, legs and face. These are also the areas of poorer circulation – the last to obtain heat from the blood (think of which parts of your body get cold first in really low temperatures). Pigment can be produced by the allele *c* only *at low temperatures*. Other parts of the cat's body are subjected to higher body temperatures and so no dark pigment is produced. As the cat gets older, these areas may darken more as circulation becomes poorer and a greater proportion of each extremity is colder. The phenotypic expression of colour is therefore influenced by the temperature of the environment.

An example of the effect of the environment on plant phenotype is seen in flower colour in hydrangeas. The acidity or alkalinity of the soil influences the colour of the flowers. Hydrangeas growing in acidic soil develop blue flowers, whereas those grown in alkaline soil develop pink flowers (Fig. 4.20c, d).

Another example of how the environment affects phenotype is when a change in temperature or pH causes a change in the shape of the active site of an enzyme, increasing or decreasing its binding with the substrate and therefore affecting its functioning (refer to Year 11 work on enzymes).

A simple example of how the environment affects gene expression can be seen in the growth of humans. Human height and infant birth weight have a genetic basis, but lack of nutrients or the presence of toxins (such as those in cigarette smoke) can restrict growth. Another well-researched example is that of the disease phenylketonuria (PKU). This is a rare genetic disease whereby the amino acid phenylalanine builds up in the body and results in symptoms that become increasingly severe as time passes. These symptoms range from behavioural and emotional problems through to developmental delays such as stunted growth, seizures and brain damage. Early intervention with a diet low in proteins that contain phenylalanine can affect gene expression, slowing down the onset of the disease and keeping the symptoms at bay.

FIGURE 4.20 The effect of environment on phenotype: **a** young Siamese cat; **b** dark-tipped older Siamese cat; **c** blue hydrangea in acidic soil; **d** pink hydrangea in alkaline soil

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Regulation of genes for phenotypic expression

Gene expression can be understood in terms of the switching on and off of genes, as needed. Investigations into gene expression have shown that gene expression may be regulated at various stages of DNA transcription into RNA and its subsequent translation into proteins, as discussed below. An interesting part of this research is that gene expression may also be affected by changes in the cellular environment.

Gene regulation by modifying DNA for transcription

Initiation of transcription seems to be determined by how densely DNA is bound to histones and this in turn affects gene expression. Epigenetics explores how chemical modifications to either the histones or the chemical structure of DNA (without changing the base sequence) may affect the density of DNA binding and, in turn, affect the switching on or off of genes. Regions of DNA that are tightly bound are inaccessible to RNA polymerase, the enzyme that starts transcription. Chemical changes such as methylation (adding a methyl group) and acetylation (adding an acetyl group) to non-base parts of DNA or to histones has been found to affect transcription. Generally, DNA methylation represses transcription, and loss of methylation activates genes. This is because methylation appears to increase the density of binding between DNA and histones, acting as a 'muffler' in silencing gene expression, whereas adding an acetyl group seems to have the reverse effect, making DNA accessible to RNA polymerase and thereby promoting transcription (Fig. 4.21). Methylation and/or acetylation of histones may have similar effects on DNA binding and gene expression. This raises the question of whether these epigenetic changes can be inherited from one generation to the next. Research shows that most epigenetic markers are wiped clear at the start of embryonic development, but some are not, leading to interesting current research into whether changes in the environment that cause 'tagging' of DNA with epigenetic markers can cause heritable variations.

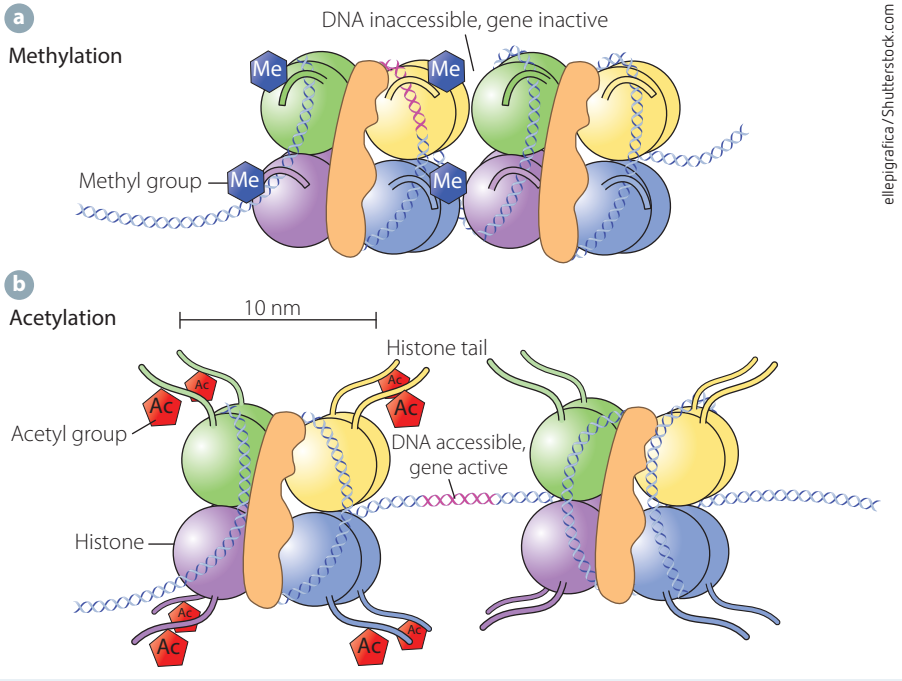

FIGURE 4.21 Chemical changes affect transcription and gene expression: **a** methylation of DNA makes it tightly packed, which 'silences' the genes because they cannot be accessed by transcription factors and be expressed; **b** acetylation of DNA promotes transcription.

Cancer was the first human disease to be linked to epigenetics. In 1983, researchers investigated the chemical structure of DNA in diseased tissue from patients with colorectal cancer and compared it with DNA in normal tissue from the same patients. The DNA in the cancerous cells of these patients had less methylation than DNA in normal tissue from the same patients. DNA that is not being transcribed in a

cell is normally methylated (it has a methyl group added to its cysteine base) and this turns off the genes. A decrease in DNA methylation can cause abnormally high gene activity – this is typical of cancer cells, which divide uncontrollably. Too much methylation can also be harmful, by not allowing the action of protective tumour suppressor genes. This has led to a flourish in research to investigate links between epigenetics and disease.

Gene regulation during the transcription process

Elongation of RNA and the termination of transcription are other steps in the process of transcribing DNA to mRNA where regulation of gene expression may occur. Protein factors influence whether mRNA elongation continues or terminates, and production of these factors relies on other genes. If the production or binding of transcription factors is affected, gene expression will be affected. These proteins that regulate transcription are also produced by genes, so this means that some genes encode their own functioning.

Post-transcription gene regulation – modifying and processing RNA

Processing of mRNA before it leaves the nucleus is thought to be one of the most common forms of gene regulation in eukaryotes. RNA processing and modification may involve:

- alternative splicing (removing introns) – as described on page 124 (Fig. 4.14)
- regulating the length of time for which mRNA remains stable. The longer an mRNA molecule lasts, the more protein will be translated. If the mRNA degrades more quickly, less protein will be made.

Small non-coding RNA molecules called microRNA (miRNA) can silence mRNA after transcription. The miRNA molecules may pair with and silence mRNA by:

- cutting RNA into two pieces
- making RNA unstable by shortening its poly(A) tail
- interfering with the translation of RNA on ribosomes.

Post-translation gene regulation – modifying proteins

After translation, the protein product made from mRNA may be inactive until a chemical is added, or it may be active until a chemical group is removed, regulating phenotypic expression.

A summary of stages at which gene expression in eukaryotes may be regulated is shown in Figure 4.22.

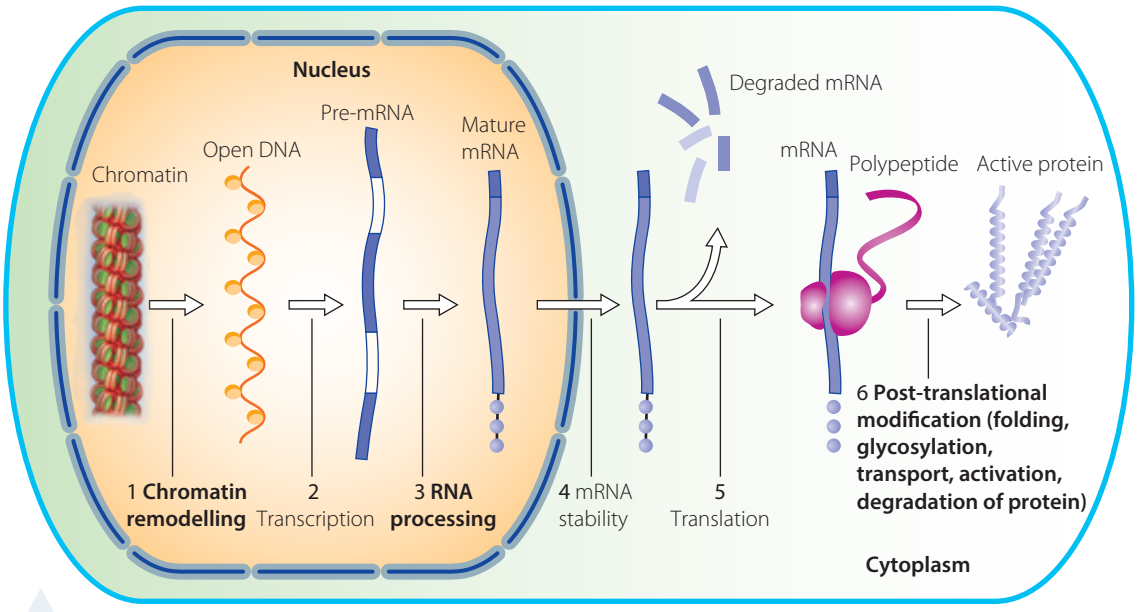

FIGURE 4.22 Regulation of gene expression in eukaryotes

An operon is a set of genes that is transcribed under the control of an operator gene. It includes the structural genes, an operator gene and a regulatory gene.

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The regulation of gene expression in prokaryotes differs from that of eukaryotes, because prokaryotes do not have introns. Bacterial gene regulation has been studied in detail by looking at the LAC operon. The LAC operon relies on the fact that some bacteria use lactose for sustenance, but most bacteria use glucose. If no glucose is present, bacteria turn on a gene that allows them to turn lactose into glucose.

INVESTIGATION 4.3

A secondary-source and practical investigation into the effects of environment on gene expression and resulting phenotype

You may do either investigation A or B, or both. Or you might wish to conduct investigation A and design (but not conduct) investigation B. Conducting investigation B is an advanced option and your choice will depend on the resources available in your school, your areas of interest and investigative skills, and the time and resources available to your teacher. Conducting investigation B may be part of a depth study.

INVESTIGATION A

You are to plan and conduct an investigation to demonstrate the effect of environment on the phenotype of 'genetic barley'.

BACKGROUND INFORMATION

Both genotype and environment may influence the phenotype of an organism. For an investigation to be valid, there can be only one variable – in this case, either genotype or environment. Because this investigation explores the effect of environment on phenotype, the genotypes of the organisms used in this investigation must be kept the same (genotype is a controlled variable). This will ensure that any change evident in phenotype has been influenced by the change made to the environment. Genetic barley is an F_1 hybrid where all seedlings are genetically similar. This is an example of a plant that may be used to allow you to determine the effects of environment on the phenotype of a plant. There are others that may be used, depending on your preferences and those of your teacher, and on the availability of equipment.

METHOD

1. Identify and clearly express the problem.
2. Form a clear hypothesis.
3. Choose appropriate equipment and conduct a risk and safety assessment.
4. Use a logical scientific method – select a suitable strategy to investigate the question, one that allows valid and reliable results to be collected.
5. Identify controls and variables and use an adequate sample size.
6. Measure, observe and record results in accessible and recognisable forms; carry out repeat trials as appropriate.
7. Use appropriate data collection techniques.
8. Assess the accuracy of your measurements and calculations, and the relative importance of the data gathered.
9. Draw valid conclusions from the data – identify trends and patterns, justify inferences and conclusions, and generate plausible explanations related to your observations and to contradictions in data and information.

Refer to Chapter 1 to revise how to formulate a hypothesis, plan a valid investigation and communicate your findings.

RESULTS

Use appropriate methods to analyse, process and present your results.
Present your investigation in the form of a scientific report.

»

DISCUSSION

1 Conduct a secondary-source investigation to find results of other similar investigations. Assess whether they support or refute the findings of your own practical investigation, and how they may add to your findings.
Evaluate the methodology that you used and describe how you could improve your investigation plan.

Revise experimental error in Chapter 1, pages 19–21.

2 Explain what may have given rise to the patterns, trends and/or relationships in your discussion. Use secondary resources to help you explain the science behind your findings.

3 Justify why you could/could not make a valid deduction as to whether the changes in phenotype that you observed and measured were due to the effect of the environment and were not due to genetic differences.

4 Assess the reliability and accuracy of your investigation, discussing any errors that may have arisen due to experimental error.

5 Evaluate the methodology that you used in your investigation and use secondary sources to help you suggest improvements to this method. Explain how you would modify your investigation plan to improve it for a future investigation of this type.

6 As a whole class, discuss the advantages and disadvantages of conducting this investigation.

CONCLUSION

Draw a valid conclusion based on your results. (There should be no inferences or explanations in your conclusion.)

INVESTIGATION B (ADVANCED)

This part is a secondary-source and practical investigation into the effects of environment on gene expression and resulting phenotype in prokaryotes.

Weblink
Jacob-Monod hypothesis for gene regulation

1 Secondary source investigation – research theoretical background information.
Go to the weblink and read the Jacob-Monod hypothesis for gene regulation.

2 Practical investigation – research practical background information on experimental design for an investigation of this kind.

In the weblink *Gene induction*, read the experimental design to investigate the expression of a gene in the bacterium *E. coli* that is switched on in the presence of lactose, noting how both qualitative and quantitative data can be collected for the investigation.

Weblink
Gene induction

3 Design a practical investigation (optional for advanced study).

Using your understanding of the practical methodology, as well as your knowledge of how the β-galactose structural, regulator and operator genes work together, design an investigation to find out what would happen to the production of β-galactosidase if one of the genes was mutated or a different sugar was introduced. To help you decide on your investigation question and design, follow these steps:

Weblink
LAC operon
Explain how a repressor protein prevents the transcription of RNA and how the presence of lactose in the environment affects this.

a Create a chart:

What do we know?

What do we think we know?

What do we want to find out?

Think of a specific question, such as:

- What would happen if the regulator gene was deleted by a mutation?
- Does glucose have a similar effect on the enzyme to that of lactose?

b State the hypothesis of the original experiment described in step 2. Create your own (revised) hypothesis for what you wish to find out.

c Design an investigation to test your hypothesis.

Using your understanding of the procedure above and the functioning of the structural, regulator and operator genes for β-galactose manufacture, design a practical investigation that could be conducted using

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» the equipment available in a school. Research methods that may be applied using simpler equipment, if necessary.

d Conduct the experiment (if your school has the resources).

e Use secondary sources to find results of other similar investigations and assess whether they support or refute the findings of your primary investigation, or to add to your findings. Use your secondary-source investigation to research answers to the questions that have arisen as a result of your investigation, to find out whether or not your experimental results are as expected in the literature, and to explain the science behind your findings. Evaluate the methodology that you used and describe how you could improve on your investigation plan.

f As a class, formulate new, revised hypotheses that could be researched further. (You do not need to conduct this further research.)

INVESTIGATION 4.4

Secondary source investigation to assess how genes and environment affect gene expression

Literacy

Critical and creative thinking

Personal and social capability

You are to research, analyse and evaluate the available literature to assess how genes and the environment affect phenotypic expression. You will need to identify and research the effect of both genes and the environment on at least one phenotypic trait.

Your research may lead you to studies that explore epigenetics as well. If so, take care to evaluate the validity, reliability and accuracy of your sources and use your critical thinking skills to draw an evidence-based conclusion.

Once you have read widely, formulate a research question or hypothesis. When gathering information, bring together the results of different studies, pointing out where researchers agree or disagree, and where significant questions remain. Draw your own conclusions, presenting your perspective as an evidence-based argument, and identify any gaps in the research to indicate directions for future research.

The difference between a research question and a research hypothesis depends on how much is known in the field. If a large amount of literature is already available and you are able to make a prediction about results, formulate a hypothesis. If little is known and you need to explore to find information, use a research question. Remember to group the literature according to common themes and then explain the link between the research question/hypothesis and the literature reviewed.

Review proposing a research question and formulating a hypothesis in Chapter 1, pages 11–12.

Follow the three rules of effective research:

- variety (use a range of key words and multiple sources)
- reliability (use information from trusted sources)
- consistency (ensure information is relevant to the topic).

At the end, pair up so that you can peer assess a literature review written by another member of your class. As a class, determine criteria for peer assessment before you begin writing your literature reviews.

PART A: CRITIQUE A LITERATURE REVIEW

Read how to write a literature review (Chapter 1, page 9) and then read the example of a simple literature review at the weblink provided here. Critique the example on the weblink, outlining three strong points and two areas where it could be improved.

Weblink
Literature review: Is the ability to dance determined by genes?

»

PART B: CONDUCT YOUR LITERATURE REVIEW

1. Do wide reading to define the topic and make some notes on opposing points of view.
2. Formulate (write) a research question or hypothesis to narrow down your secondary-source investigation. (See page 8 on proposing a hypothesis or research question.) Write a list of key words that you could use to find answers to your research question.
3. Find relevant articles using databases, Internet search engines and library catalogues. Outline opposing points of view in the research and critically analyse the validity of views expressed in your sources. (See page 10 on evaluating sources.) Draw a conclusion to your research question through an evidence-based argument. Acknowledge your sources using an accepted referencing style.
4. Communicate your information clearly and accurately. Use the correct structure for writing a literature review: introduction, body and conclusion. See page 9 for more details on how to structure a literature review and what type of information to include in each part.

   Remember to use correct scientific terms and keep the language at a level that is suitable for Year 12 students to peer review your work.
5. Draw your own conclusions about the research, based on your findings. Express your perspective on the strengths and weaknesses of the research you are reviewing and use your findings to support your judgement.

   Your literature review should be 400–800 words. This may vary, depending on your research question and the depth of your research. Seek permission from your teacher before making your literature review longer than this. Remember: it is quality, not quantity, that counts.
6. Peer review at least one literature review written by someone else in your class, using criteria drawn up by the class. See page 4 in Chapter 1 for a reminder of what to look for when conducting a peer review.

KEY CONCEPTS

- Gene expression is the switching on and off of genes to make the required proteins and other end products in particular cell types.
- Phenotypic expression is the result of gene expression – the structure, physiology and behaviour of an individual as a result of genes that have been expressed.
- Some variations within a population are due to the influence of the environment, rather than having a genetic (DNA sequence) basis.
- Identical twins have identical genotypes, and therefore any phenotypic differences can be attributed to environmental influences.
- An example of variation brought about by the environment is the difference in colour of flowers in hydrangeas (dependent on pH of the soil).
- Chemical modifications of DNA that do not involve a change in the sequence of nucleotides are termed 'epigenetic' modifications. They may be the mechanism by which some environmental factors bring about variation. The result is a change in phenotype without a change in genotype.
- Epigenetic changes show links to disease, including cancers and metabolic diseases.

CHECK YOUR UNDERSTANDING 4.3

1. Using examples, explain how:
   - **a** genes affect phenotype
   - **b** the environment may affect phenotype in a manner that is not heritable
   - **c** the environment may affect phenotype in a manner that is heritable.
2. Identify reasons why cells do not express all the genes in their genomes.
3. Outline how the packaging of DNA affects gene expression.
4. Compare the effects of methylation and acetylation of DNA on gene expression.
5. Explain, using an example, how epigenetics may account for a phenotypic change.
6. How do scientists account for the fact that humans have fewer genes than the number of types of proteins in cells?

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4.4 The structure and function of proteins in living things

Proteins are the most abundant organic molecules in cells. They are responsible for forming the basic structure of cells and for carrying out all the work in cells, by controlling all chemical reactions.

Revise your Year 11 work on protein structure.

Worksheet
Protein revision

Structure of proteins

Proteins are made up of one or more long chains of nitrogen-containing amino acids. Each chain is called a polypeptide. Each protein within an organism is folded into a particular shape that is crucial to its functioning. Proteins bind with other molecules to carry out their functions, and so their shape and chemical properties (such as electrical charge and attraction to water) allow the proteins to function in a particular way.

Chemical structure of proteins

Proteins contain the chemical elements carbon, hydrogen, oxygen and nitrogen, and sometimes sulfur. These elements combine to form amino acids, which are the building blocks of proteins. There are about 20 amino acids; they can be put together in chains of up to 300 amino acids. The amino acids in each linear sequence or polypeptide chain are held together by chemical bonds (forces of attraction) known as **peptide bonds**. *One or more polypeptides* can be twisted together into a particular shape, resulting in the overall structure of a *protein*. The *sequence and arrangement of the amino acids* determines the type of protein, in the same way that sequences of the letters of the alphabet can be used to make words and then sentences.

If a single chain of amino acids is longer than 40-50 amino acids and folded in a specific manner, it is termed a *protein*. If the chain is shorter than 40-50 amino acids and combines with other chains to fold into a functional protein, it is termed a *polypeptide*.

Models of the protein insulin can be seen on page 120 (Fig. 4.9).

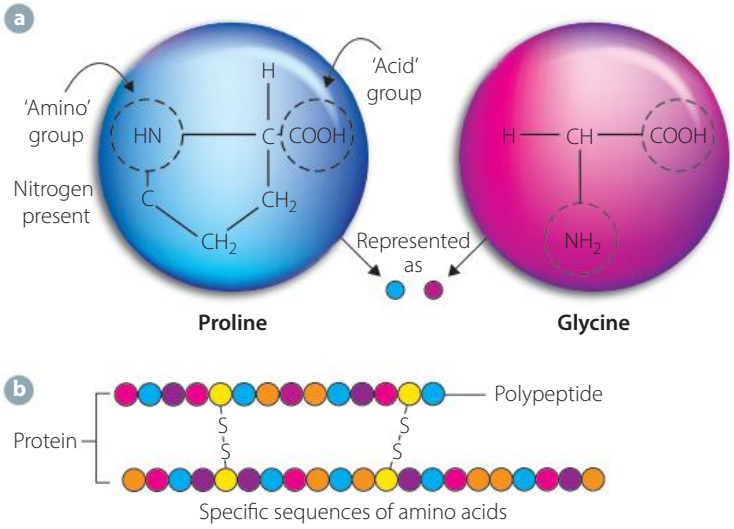

FIGURE 4.23 Protein structures: **a** structural formula of two amino acids, proline and glycine. The hydrocarbon chains of amino acids differ in length and are known as the 'R' group; **b** two polypeptide chains of amino acids held together by peptide bonds (chain), with chains held together by sulfide bonds, making up a protein (e.g. insulin)

Physical structure of proteins

The structure of proteins can be described at four levels: primary, secondary, tertiary and quaternary (Fig. 4.24).

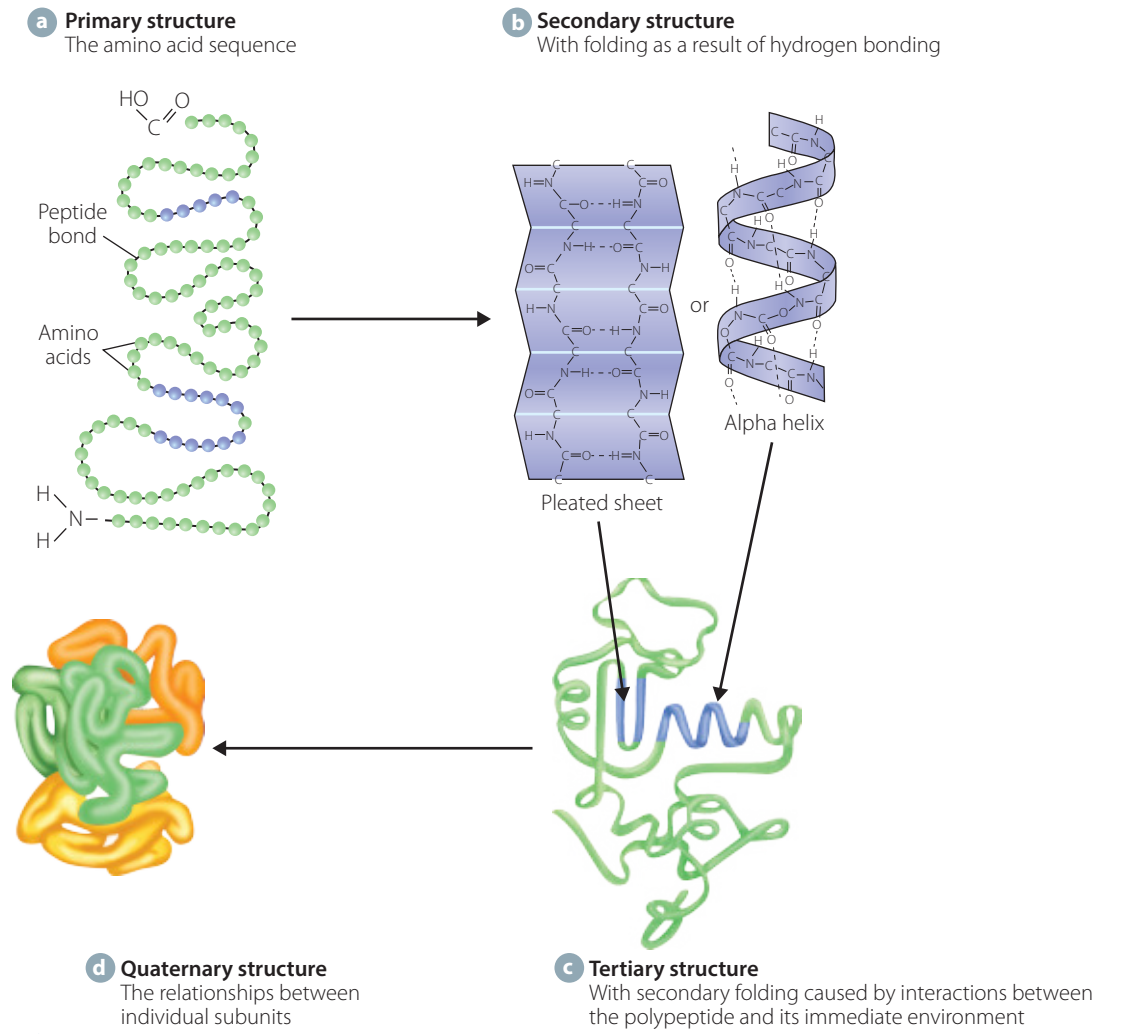

FIGURE 4.24 The four levels of protein structure: **a** primary, **b** secondary, **c** tertiary and **d** quaternary

The basic structure of a protein is a polymer of amino acids, arranged in linear chains or polypeptides, and is termed its **primary structure**. However, it is the shape of the protein, not merely the amino acid sequence, that determines its function. Proteins have a hierarchy of folding that gives them their specific shapes.

Proteins made up of a single polypeptide chain have primary, secondary and tertiary structure. Proteins made up of more than one polypeptide chain have quaternary structure.

The **secondary structure** is the three-dimensional arrangement of the polypeptide chain. The secondary structure forms as a result of the amino acid chain becoming linked by hydrogen bonds, either twisting the polypeptide into a spiral (alpha helix) typical of a fibrous protein, or folding it into a pleated sheet rather than a spiral, also held together by hydrogen bonds.

Further folding leads to the **tertiary structure** that is seen in more complex proteins such as globular proteins. Certain forces of attraction between alpha helices and pleated sheets (such as disulfide bonds) cause the polypeptide to fold into a more complex three-dimensional shape.

Quaternary protein structure occurs in proteins that are made up of two or more polypeptide chains that link to create a more complex three-dimensional structure.

A single protein molecule may contain more than one type of protein structure. For example, the silk of a spider contains pleated sheets joined by less ordered alpha helices.

Some proteins, called conjugated proteins, are linked to a non-protein part called a **cofactor**. If the cofactor is *tightly bound*, it is termed a **prosthetic group** and may be organic or an inorganic metallic ion.

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The blood pigment haemoglobin contains inorganic iron as its prosthetic group. If a cofactor is *loosely bound* to an enzyme, it is known as a **coenzyme** (often an organic molecule such as a vitamin).

The primary structure of a protein (the linear amino acid sequence) determines the secondary and tertiary structure of the protein, particularly those amino acids that are involved in forming bonds to create the spiral or pleated sheets. If the incorrect amino acid is inserted in a polypeptide chain, this may lead to a change in the bonding properties of an essential part of the protein and therefore change its secondary, tertiary and quaternary structures, which could change the functioning of the protein. The resulting protein often functions less effectively and the change may even be lethal. For example, in the inherited red blood cell disease known as sickle cell anaemia, a single amino acid change in the protein haemoglobin is the cause. The amino acid glutamate is replaced by a valine. This distorts the shape of the haemoglobin protein (changes it to long fibres) and affects its ability to transport oxygen. This is not a lethal mutation, unless inherited in both maternal and paternal copies of the gene. Occasionally, a change in an amino acid may lead to improved functioning of a protein. Changes such as these are the cause of random variation in a population that is subject to natural selection. For example, if a change in an enzyme has no effect on the organism at the natural ambient temperature, but allows it to function more efficiently in high temperatures, then an environmental change such as global warming may result in that individual surviving and passing on its gene to the next generation.

Sickle cell anaemia is dealt with in more detail in chapters 5 and 7.

Types of proteins in cells

As you have learned, the structure and shape of proteins determine their functions, which range from structural support to cell communication, movement, defence against diseases and cell biochemistry. Different types of tissues contain different proteins, coded by DNA that is switched on during cell differentiation.

Fibrous proteins form structural components of cells and tissues; together with water, they form the basic structure of protoplasm (the cytoskeleton). Fibrous proteins are long and insoluble in water. Collagen is an example of a long, stringy protein that is coiled and very strong (Fig. 4.25a). It is commonly found in skin, along with another fibrous protein, elastin. These proteins are also present in ligaments and tendons. Keratin is a fibrous protein in hair and nails.

Globular proteins are usually spherical in shape and are compact and soluble in water. They are often transport proteins, such as haemoglobin in the blood (Fig 4.25b). Immunoglobulins (antibodies), hormones and enzymes are other examples of globular proteins.

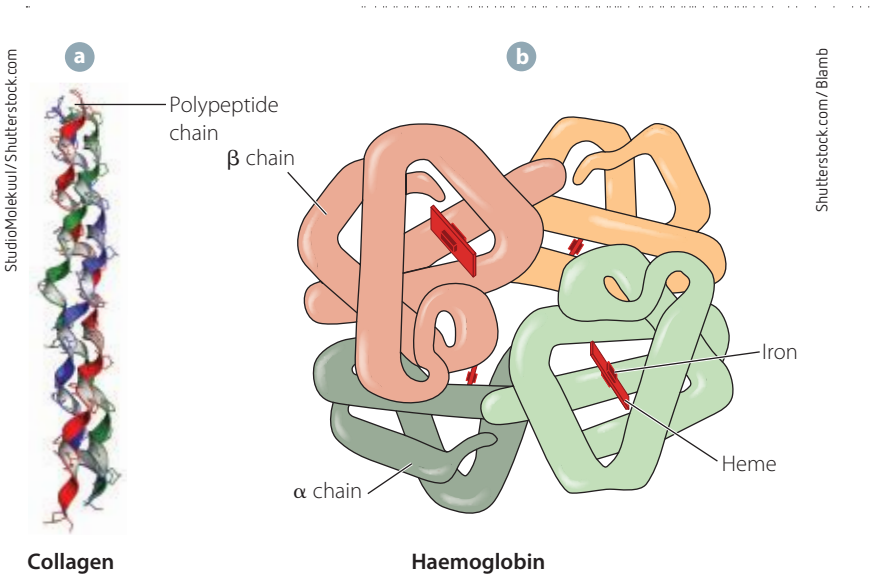

FIGURE 4.25 Fibrous and globular proteins: **a** collagen, a fibrous protein; **b** haemoglobin, a globular protein

KEY CONCEPTS

- Proteins are macromolecules (polymers) made up of one or more polypeptide chains that are formed by sequences of amino acids linked by peptide bonds.
- Proteins contain the chemical elements nitrogen, carbon, hydrogen, oxygen and sometimes sulfur.
- Proteins have a primary structure (sequence of amino acids in polypeptide chains), secondary structure (polypeptide chain arranged in a helix or pleats), tertiary structure (3D shape formed by folding the secondary structure) and quaternary structure (two or more polypeptide chains combined).
- The sequence of amino acids in a protein is responsible for the overall bonding, folding and 3D shape of a protein, which determines its ability to function. A change in one or more amino acids in the sequence has the potential to alter the entire structure of a protein.
- There are three main types of protein: fibrous, globular and conjugated.

CHECK YOUR UNDERSTANDING 4.4a

1. Name four structural proteins in cells and state the function of each.
2. Identify three types of functional proteins in cells and, using an example of each, explain how they function.
3. Name the types of bonds that form between amino acids in a polypeptide and explain how they form.
4. Outline the primary, secondary and tertiary structure of a protein.
5. Using examples, explain how an amino acid substitution in a polypeptide may affect its functioning.

Functions of proteins

The biological properties of proteins depend on the interactions of the proteins with each other and with other molecules. For example, enzymes bind with substrates to catalyse reactions, regulatory proteins control DNA replication and turn on genes, antibodies bind with pathogens, and hormones bind with receptors on target cells.

The tertiary structure and three-dimensional shape of a protein determines its ability to bind tightly and specifically with these molecules, and therefore determines its ability to function effectively.

Proteins are reusable, and reactions between them and their binding molecules (called **ligands**) are reversible.

Proteins can be classified into categories according to their functions. In the section that follows, proteins are grouped into five categories according to function. Note that each category may be subdivided into further groups – see the weblink for proteins grouped into nine categories, according to function.

Weblink
Types of proteins
Explore the different types of proteins and their function.

Structural proteins – support and movement

Structural proteins are often fibrous and stringy (such as collagen and elastin), and found in connective tissues such as skin, cartilage, bone, tendons and ligaments. Structural proteins also make up shells in invertebrates, and are in hair, nails and hooves in vertebrates.

Structural proteins such as tubulin (in microtubules) are responsible for forming the cytoskeleton, which maintains the shape of cells. Microtubules also allow movement in cells. For example, cilia and flagella in cells move as microtubules slide along each other, and a similar action causes spindle fibres to contract.

Contractile proteins also occur in cells, allowing movement to occur. For example, in muscle, the protein actin slides along another protein, myosin, to allow the muscle to contract (Fig. 4.26). Actin is also found in microfilaments in cells and these are responsible for contraction of the cytoplasm, allowing the cell membrane to pinch off during cytokinesis in animal cells, and the crawling movement of protists such as amoeba.

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Enzymes – control of biochemical reactions

Enzymes are protein molecules involved in all biochemical aspects of cellular metabolism. (See Year 11 course work.) They catalyse reactions such as those in the chemical respiration pathway and in digestion. The shape of an enzyme's active site determines its binding specificity and therefore its ability to function. Enzymes are particularly important in gene functioning, replicating, repairing and transcribing DNA to make new proteins.

Review *Biology in Focus Year 11,* Section 3.3 on enzymes.

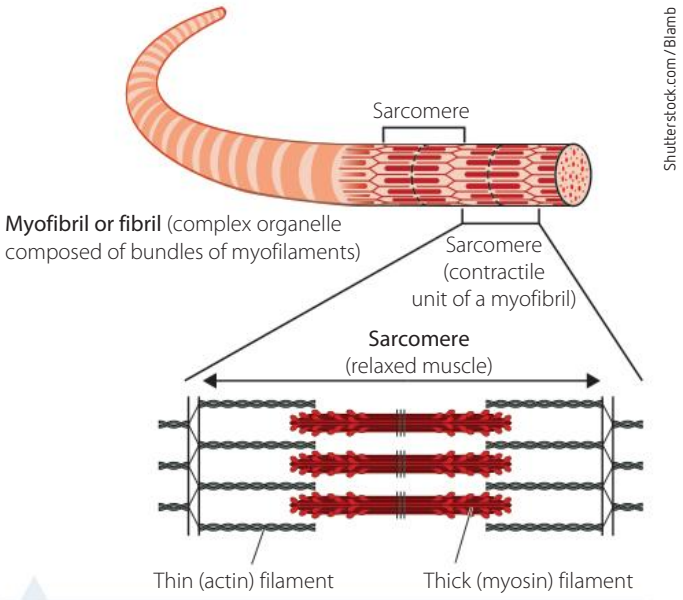

FIGURE 4.26 Thick myosin filaments in muscle slide along thin actin filaments, allowing the muscle to contract.

Proteins for cell communication, cell signalling and biological recognition

Cells communicate by means of chemical signals. Some proteins embedded in membranes form channels to carry substances that are essential for cell functioning between cells and the environment (Fig. 4.27). (Revise your Year 11 work on the structure and functions of cell membranes.) For example, nerve cells and cells in kidney tubules rely on proteins embedded in the cell membrane to act as a sodium pump, regulating the intake and output of sodium ions so that the cell can function.

See *Biology in Focus Year 11,* Chapter 3, on the structure of the cell membrane.

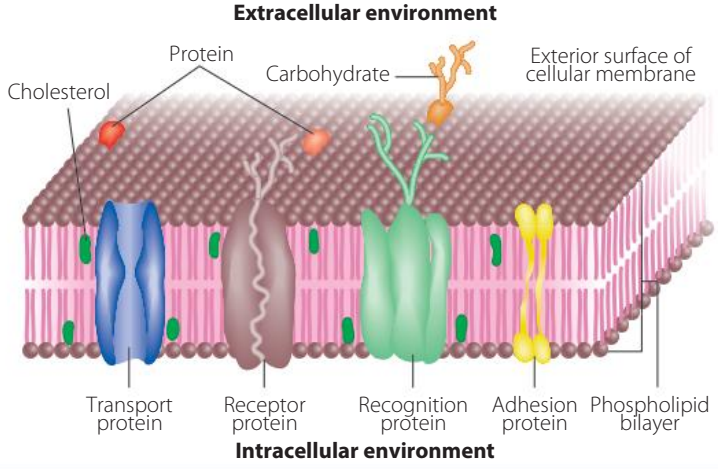

FIGURE 4.27 A view of the fluid mosaic model of part of the cell membrane, showing embedded and surface proteins.

Cell signalling and biological recognition

Some proteins, such as hormones and neurotransmitters, act as chemical messengers between cells. They communicate messages to a cell about the environment around it and trigger responses in the target cell's functioning. Receptor proteins in the membranes of cells are responsible for receiving these signals. Biological recognition between chemical messengers and their target cells is essential. Receptor proteins must therefore have a shape that is exactly reciprocal to the shape of the molecule with which it binds.

Some receptors on the surfaces of cells are genetically determined. Some of these receptors act as markers that allow a body to recognise its own cells and the cells of foreign invaders (pathogens) that need to be destroyed. This helps the body to fight disease. Antibodies are defence proteins that may be attached to cell membranes or floating freely in body fluids. Antibodies (also called immunoglobulins) recognise foreign invaders by the different proteins on the surfaces of their cells (called antigens); the antibodies bind with these antigens to signal to other defence cells in the body that the invading antigens need to be destroyed (See Chapter 12 for more details.)

Antibodies are dealt with in more detail in Chapter 11.

Transport and storage proteins

Some proteins bind to and carry or store chemicals in the body. These are termed ligand-binding proteins and must be able to bind easily with the chemical (ligand) and also release it when and where it is needed. An example is haemoglobin, which has an affinity for (attraction to) oxygen, particularly when oxygen is present in high concentrations. Haemoglobin loses its affinity for oxygen in areas where there is a low oxygen concentration (and high carbon dioxide concentration), such as in active tissues. The protein changes shape when the oxygen level changes, and this adaptive ability of the protein is essential to its functioning.

Some proteins store chemicals for use by the organism – for example, ferritin stores iron, while albumin (in egg whites) and casein (in milk) store amino acids.

Weblink
How a firefly's tail makes light
Identify the series of proteins involved in generating light, and classify each into a group according to its function.

Sensory proteins – responding to stimuli

Some proteins change their shape or biochemical activity in response to stimuli (changes in the environment). For example, opsins are proteins in the retina (innermost layer) of the eye that detect light. When light is absorbed by cells in the retina, these proteins undergo a change in molecular arrangement and start a series of reactions in which light energy is transformed into electrical and chemical signals that can be interpreted by the brain.

Refer to Chapter 18, page 612 to learn more about the functioning of the protein opsin in vision.

KEY CONCEPTS

- Proteins are grouped according to their structure or functioning to make it easier to remember and compare them.
- Structural proteins form the structural and functional part of cell membranes. Some proteins in the cell membrane also function in communication and regulate the movement of substances across the cell membrane.
- Some proteins, such as myosin and actin in muscle cells, are responsible for cell motility, joining together to form filaments that allow movement.
- Storage and transport proteins bind and store or carry other molecules in cells. For example, histones package DNA in a compact form, the protein ferritin stores iron and haemoglobin carries oxygen.
- Some proteins regulate metabolic functioning – these include enzymes (chemical catalysts) and chemical messengers such as hormones and neurotransmitters.
- Proteins involved in cell recognition include antibodies, which defend against disease.

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Technology, big data and the investigation of protein structure and function

Today, we can predict expected amino sequences in proteins, as we have increased our understanding of how the millions of nucleotides in a sequence of DNA are decoded. Sequencing the nucleotides of complete *sets of genes* in organisms is a field known as **genomics**. **Proteomics** is the large-scale study of *sets of proteins* produced in an organism or biological system to find out how the proteins in a variety of cell types work together. Studies of both genomics and proteomics have made major leaps in recent years as a result of advances in computer technology. (See the weblink.)

The genome of an organism remains more or less the same in different cells over time, but the proteome is more complex. Various cell types produce different sets of proteins and these are often modified after translation. Different genes are expressed in different cell types, so there are an enormous number of proteins to be identified and studied.

Analysis of proteins has also made rapid advances over the past 30 years or so, as has analysis of DNA and gene functioning.

X-ray crystallography was first used over a century ago. Over the past 60 years or so, it has been used to study biological macromolecules (large molecules). Advances in the study of proteins were slow at first, as X-ray diffraction technology needed to evolve to produce sufficiently strong X-ray beams to provide diffraction images that could be measured. The images and methods of gathering data were sufficient for small molecules, but collecting data for large molecules such as proteins took many days or weeks.

The study of the first protein (myoglobin) whose entire structure was recorded was published in 1958. By 1971, a worldwide database, the Protein Data Bank (PDB), had been created and seven protein structures had been deposited in the PDB, all determined using single-crystal X-ray diffraction. The numbers increased slowly – by 1973, two more proteins had been deposited.

Because one protein may have between five and 50 different modified forms, each allowing the protein to perform a different function, the process of determining protein structure was slow and the goal enormous. What was needed were high-brilliance sources of X-ray radiation to speed up the crucial diffraction measurements, and a way to analyse the data quickly.

These requirements were met over the next ten to twenty years. The development of the synchrotron (a particle accelerator) brought about this change. A synchrotron emits electromagnetic radiation where charged particles can be accelerated to move at speeds close to the speed of light. The particles can be forced to change direction by a magnetic field. Synchrotron technology improved and by the 1990s, the construction of a third-generation synchrotron with a ring size of over a kilometre gave the high-resolution images required. This, together with the advent of high-performance computing and the use of recombinant methods for protein production, advanced the study of proteins. Molecular biologists were able to record and create models of the structures of thousands of proteins and bank this data in a relatively short space of time. The graph in Figure 4.28 compares the numbers of protein structures determined using synchrotron radiation (orange) and conventional sources of radiation (blue) between 1985 and 2009.

The field of combined computer science and genetics is known as **bioinformatics**. People who work in this field develop methods and software for analysing and interpreting biological data. It is an area of science that combines the disciplines of biology, computer science, mathematics, statistics and engineering.

Advances in computer technology have led to the rapid storage and manipulation of big data (extremely large volumes of data), greatly accelerating the study of proteomics. Today, the detailed analysis of protein structure to identify functions and interactions is assisted by 3D visualisation computer technology. For example, some programs allow protein structure to be 'sorted' according to the arrangement of particular chemical elements within the molecule, bonding patterns, the line-up of atoms in sequence or the overall structure and shape of the protein. This is extremely useful for exploring how changes in amino acid sequences affect protein structure. A number of proteins targeted by medical research for cancers are currently being studied in this way, using big data.

Weblink
Timeline of genomics and proteomics
Record in order ten major discoveries relating to our understanding of DNA and protein synthesis.

Weblink
Human Genome Project

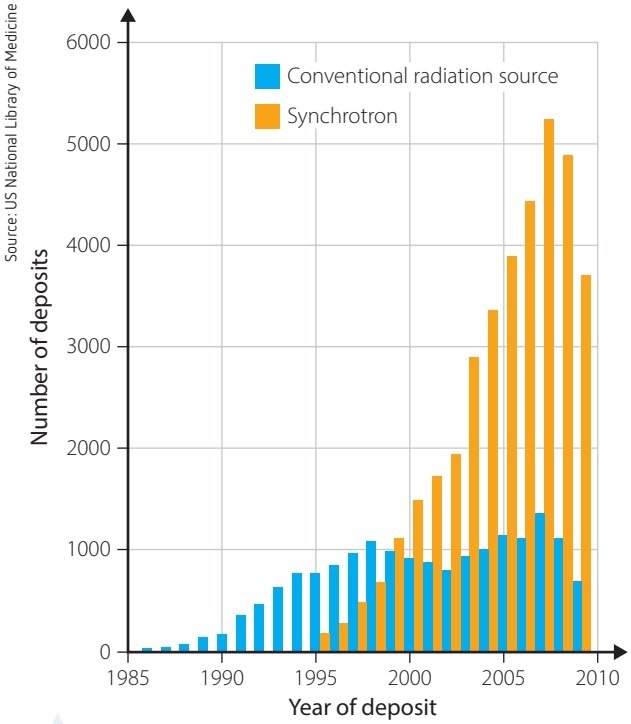

FIGURE 4.28 The number of protein structures deposited annually in the Protein Data Bank

INVESTIGATION 4.5

Information and communication technology capability

Secondary-source investigation into protein structure and function

You are required to investigate a specific protein and create a multimedia presentation of your information (Investigation part 1), after which you will research one of two specific named proteins (Investigation part 2).

PART 1: INVESTIGATING A PROTEIN OF YOUR CHOICE

Investigate the structure of a particular protein of your choice (list of suggestions in point 3 below) and how a change in its structure could affect its functioning.

1. Create a multimedia presentation of 8–10 slides, outlining information about the structure of a protein of your choice, its functions and its potential applications.
2. Find a video clip of the protein functioning, as well as a weblink to a computer simulation program that allows visualisation of the protein in three dimensions. Insert the sites as hyperlinks in your multimedia presentation.
3. Outline the impact that a change in the structure of this protein may have on its functioning. Some examples of proteins that you may wish to select for your research are: motor protein kinesin; proteins involved in DNA replication such as bacterial DNA gyrase; reverse transcriptase; tumour suppressor proteins such as p53; metabolic protein such as haemoglobin; proteins implicated in diseases such as cardiovascular disease, Alzheimer's disease, Parkinson's disease; proteins involved in defence against disease, such as immunoglobulins; major histocompatibility complexes (MHCs) or any protein of your choosing.

Weblink
Rotatable and zoomable 3D structure of proteins

Weblink
Cn3D
A visualisation tool for biomolecular structures, sequences and sequence alignments.

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» Some weblinks are provided for you to begin your research. You need to find similar sites to complete the research on your selected protein.

PART 2: HAEMOGLOBIN OR CYTOCHROME C

Research the importance of the amino acid sequence in a haemoglobin molecule or in cytochrome C and how these molecules may be used for relatedness studies in evolution. Use one of these molecules to explain the importance of DNA transcription and translation being accurate when creating the amino acid sequence of the polypeptides that make up the selected protein.

PART 3: BIOINFORMATICS (OPTIONAL – ADVANCED)

1 Do research to answer the following questions.
   - **a** What is bioinformatics?
   - **b** What kind of work does a molecular diagnostics researcher do?

2 Explain how bioinformatics may be used in the work of a molecular diagnostics researcher.

OR

Write a job advertisement for a molecular diagnostician, outlining the knowledge and skills the person would need to have, to apply for this position.

Weblink
3D visualisation of the structure of the p53 protein

Weblink
p53 and how it functions in tumour suppression

Weblink
TED talk using data to create computer-generated 3D images

Personal and social capability

Work and enterprise

KEY CONCEPTS

- The study of a set of proteins in an organism is called proteomics.
- Bioinformatics is a field of study that involves developing computer technology including software tools for understanding and visualising enormous amounts of biological data. It is a combination of biology, computer science, mathematics and engineering.

CHECK YOUR UNDERSTANDING 4.4b

1 Construct a table like the one below to compare the main functional categories of proteins in cells.

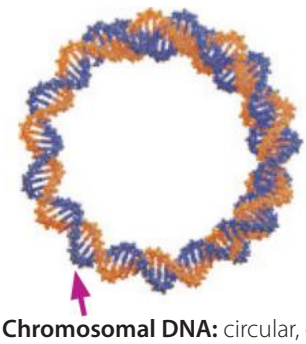
CATEGORY OF PROTEIN	FUNCTION	DESCRIPTION	EXAMPLE
Structural	Support		
	Movement		
Regulating metabolic functioning	Enzymes		
	Hormones		
Cell communication	Signalling		
	Biological recognition		
Sensory proteins	Response to stimuli		
Storage and transport	Storage		
	Transport		

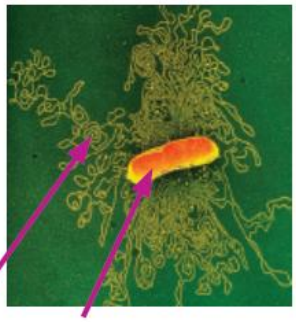
2 Distinguish between proteomics, genomics, big data and bioinformatics.

4 CHAPTER SUMMARY

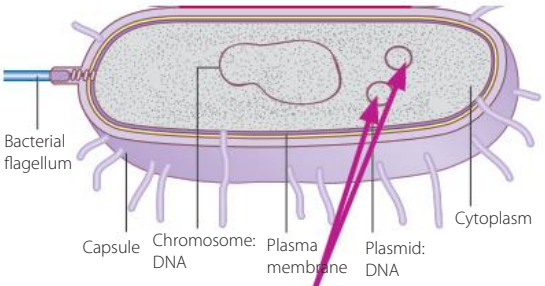
DNA and polypeptide synthesis: Why is polypeptide synthesis so important?

DNA IN PROKARYOTES



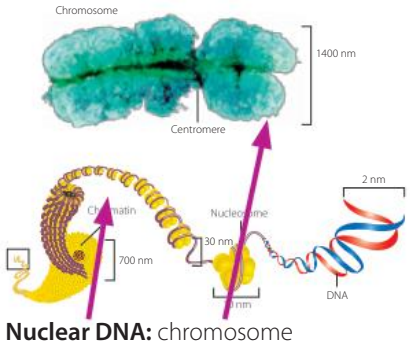


Chromosomal DNA: circular, double-stranded and supercoiled into a nucleoid



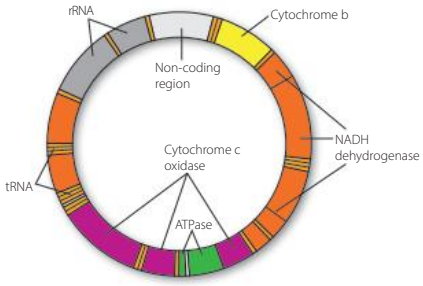
Non-chromosomal DNA: small rings of non-chromosomal DNA

DNA IN EUKARYOTES



Nuclear DNA: chromosome

Nuclear DNA is linear DNA packaged around histones (to form nucleosomes), which play a role in regulating gene expression.



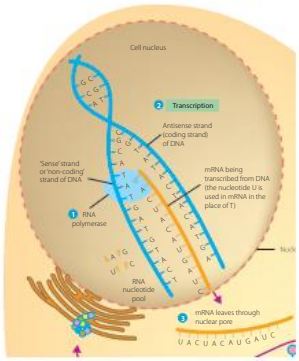
Non-nuclear DNA: mitochondrial DNA

mtDNA occurs in mitochondria in the cytoplasm, is inherited down the maternal line and mutates at a higher rate than nuclear DNA.

POLYPEPTIDE SYNTHESIS

Step 1 — DNA codes protein synthesis — **Step 2**

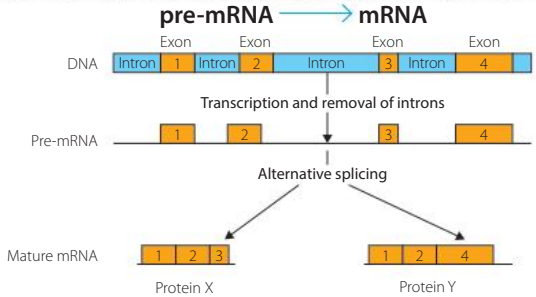
DNA → RNA



Transcription

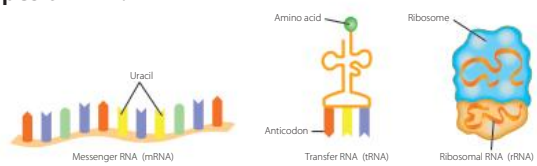
Instructions copied in a coded form from DNA, carried by mRNA into the cytoplasm

pre-mRNA → mRNA



Introns are spliced out (regulate gene expression).

Types of RNA:



mRNA **tRNA** **rRNA**

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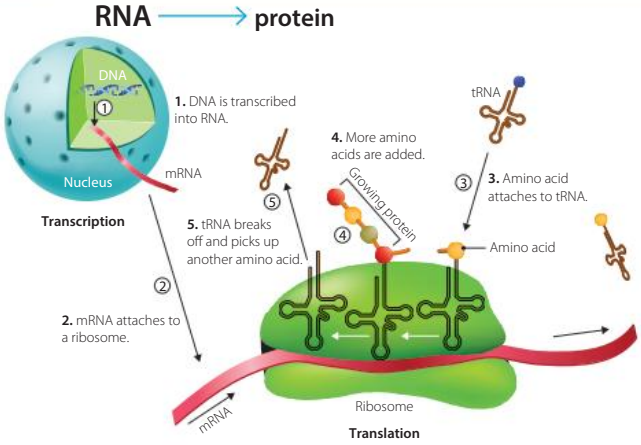
POLYPEPTIDE SYNTHESIS

Step 3 **Translation into polypeptide chains**



Translation

mRNA is 'read' by ribosomes and translated into polypeptides, with the help of tRNA.

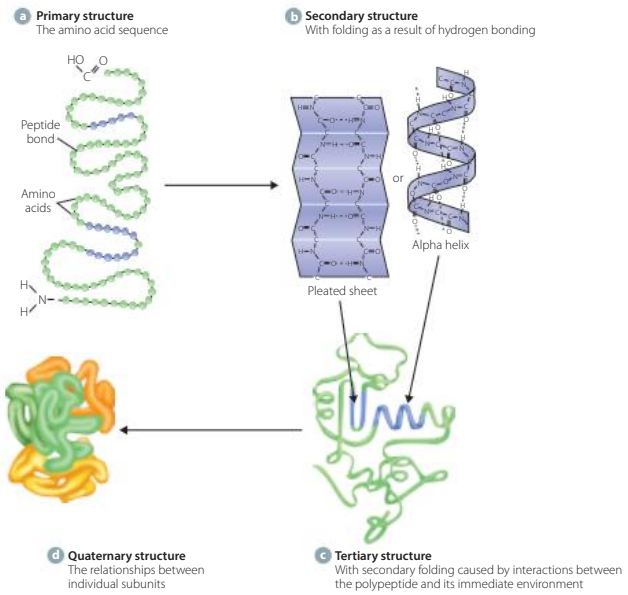
THE ENVIRONMENT CAN AFFECT PHENOTYPIC EXPRESSION

Gene expression is the switching on and off of genes to make the required protein end products in cells. Environment and epigenetics play a role.

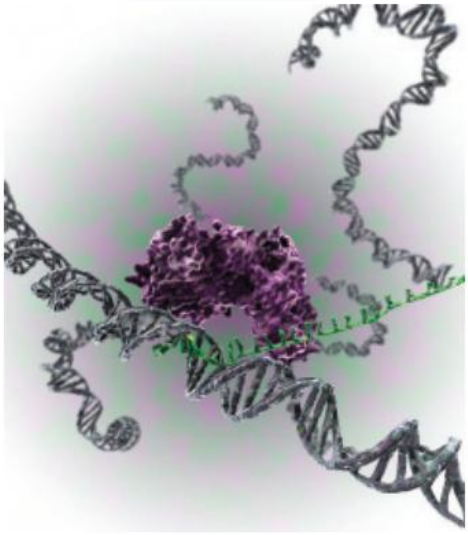
Identical twins have the same genotype. Any differences are due to environmental influence.

Hydrangea colour is affected by pH in the environment.

Proteins produced



Protein function



(DNA polymerase enzyme) depends on shape.

4 CHAPTER REVIEW QUESTIONS

Review quiz

1. Draw a diagram and annotate it to explain the structure of a eukaryotic chromosome.
2. Compare the chromosome of a eukaryote with that of a prokaryote.
3. Distinguish between the following structures:
   - **a** nucleoid and plasmid
   - **b** scaffold and histone
   - **c** nuclear DNA and non-nuclear DNA in eukaryotes.
4. Identify one similarity and two differences between a plasmid and mtDNA.
5. Explain why mtDNA is useful for evolutionary studies.
6. List the following in order of genome size, from largest to smallest: prokaryote circular DNA, eukaryote nuclear DNA, yeast DNA, mtDNA.
7. What are histones? Describe their role in the nucleus of eukaryotic cells and in regulating gene expression.
8. Using diagrams, explain how DNA is packaged in a prokaryotic cell.
9. Define 'gene'. Explain why the definition has changed over time.
10. Give three examples of how the environment affects gene expression and two examples of how the environment affects phenotype in a way that is not hereditary.
11. Draw a Venn diagram to compare DNA and RNA.
12. Use words and arrows to represent the central dogma of molecular biology.
13. Give the full name of each of the following types of nucleic acids in cells and outline the function of each:
    - **a** DNA
    - **b** mRNA
    - **c** rRNA
    - **d** tRNA
    - **e** mtDNA
    - **f** miRNA
14. Do more complex organisms have a larger number of chromosomes? Use data to justify your answer.
15. List the following in order of size, from smallest to largest:
    - **a** polypeptide, protein, amino acid, dipeptide
    - **b** nucleic acid, nucleotide, chromosome, gene.
16. Draw a diagram to represent:
    - **a** a protein molecule, and label all parts listed in Question 15a
    - **b** part of a chromosome, and label all parts listed in Question 15b.
17. Draw a diagram to represent a thymine nucleotide of DNA, labelling the three distinct chemical components. Describe any changes you would need to make to your diagram if this was a nucleotide of RNA.
18. Compare the structure of fibrous and globular proteins.
19. Distinguish between a proteome and a genome, using examples.
20. Construct a flow chart with words, arrows and diagrams, to represent:
    - **a** the process of transcription of pre-RNA from DNA
    - **b** the subsequent editing of RNA to become mature RNA
    - **c** the process of translation.
21. Create a flow chart with words and arrows to outline the sequence of events that occurs during transcription and translation. Indicate on your flow chart which steps occur inside the nucleus and which steps occur in the cytoplasm.
22. Compare transcription and translation in the DNA of prokaryotes and eukaryotes.
23. Explain the difference between coding and non-coding DNA.
24. Identify the sequence of amino acids that would result from an mRNA molecule with the sequence AUUCGUGUAGCCGGUCGA.
25. Draw the non-coding strand of DNA that would have given rise to the mRNA molecule in Question 24.
26. Discuss the importance of using models in biology, referring to the discovery of the structure of DNA by Watson and Crick as an example.
27. Construct a simplified diagram of a strand of DNA to show a sequence of nucleotides 24 bases in length, following the instructions below.
    - **a** Use a single line to represent the sugar-phosphate backbone and letters to represent the bases.
    - **b** Use the mRNA codon table in Figure 4.15 to make sure that the first triplet of bases on the DNA strand you construct codes for a start codon in mRNA and that the last triplet of bases codes for a stop codon.
28. Draw a diagram to show how the strand of DNA that you created in Question 27 would be:
    - **a** transcribed into mRNA
    - **b** translated into a polypeptide chain (use the mRNA codon table in Figure 4.15).
29. Write two changes in bases that could occur in the DNA strand that you created in Question 27 that would *not* result in a change in the sequence of amino acids. Explain why this is so.

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30 Explain the importance of twin studies in genetics.

31 Describe three ways in which gene expression can be regulated. You may use diagrams to assist your explanation.

32 Explain the importance of literature reviews in research.

33 Discuss the importance of bioinformatics in understanding both DNA and protein functioning. Use the information in Figure 4.28 to support your answer.

34 Write an extended response to discuss whether genes or the environment have a greater influence on phenotype. Support your arguments with current biological knowledge.

35 Explain what is meant by 'gene expression' and its significance in living organisms. What, then, is the purpose of polypeptide synthesis?

36 Design an investigation that a scientist could conduct to show that exons are the only coding sequences expressed in a protein.

Exam preparation

5 Genetic variation

INQUIRY QUESTION

How can the genetic similarities and differences within and between species be compared?

Students:

- conduct practical investigations to predict variations in the genotype of offspring by modelling meiosis, including the crossing over of homologous chromosomes, fertilisation and mutations (ACSBL084)
- model the formation of new combinations of genotypes produced during meiosis, including but not limited to:
  - interpreting examples of autosomal, sex-linkage, co-dominance, incomplete dominance and multiple alleles (ACSBL085) CCT
  - constructing and interpreting information and data from pedigrees and Punnett squares
- collect, record and present data to represent frequencies of characteristics in a population, in order to identify trends, patterns, relationships and limitations in data, for example: ICT N
  - examining frequency data
  - analysing single nucleotide polymorphism (SNP)

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
5.1 Secondary-source and practical investigation to model meiosis, fertilisation and mutations
5.2 Practical investigation to gather data and construct a family pedigree chart
5.3 Modelling inheritance patterns
5.4 Examining frequency data in genetic studies
5.5 A secondary source investigation of how genetic similarities and differences can be compared

Worksheets
- Chromosome terminology
- Meiosis
- Modelling meiosis: template
- Mendel's laws
- Dominant and recessive traits
- Interpreting pedigrees
- Traits and information that may be used for constructing pedigrees
- Additional information on sex determination
- Sex linkage diagrams and additional information on colour blindness
- Summary of sex-linkage in humans
- Deviations from simple Mendelian ratios
- Extension: Understanding SNP data

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

Why do a high proportion of African people have the allele for sickle cell anaemia, which gives rise to a detrimental blood disorder, even though it occurs in low frequencies in other populations? If there is cystic fibrosis in my extended family, what is the percentage chance that I may have a child with cystic fibrosis? If my mother has breast cancer, am I likely to develop it too? Is prostate cancer hereditary? What is the likelihood that my child will inherit my sporting ability or my partner's musical talent, or both? These are common questions and, using mathematical models and advanced understanding of inheritance patterns, scientists are becoming better able to answer them.


Penny Tweedie/Alamy Stock Photo

FIGURE 5.1 Studying heredity patterns can enable geneticists to predict features within a family or population.

In earlier chapters you learned that, while traits are passed on as the result of accurate replication of chromosomes, variation that arises during sexual reproduction is also important in the process of natural selection for the adaptation and survival of species.

In this chapter you will explore heredity patterns and the frequencies of variations, which can be used to compare genetic similarities and differences within and between species. Population geneticists conduct such comparisons to predict, with a fair degree of accuracy, the chance of certain features being represented within a family, population or species.

Weblink
Cultural kinship
Understanding the concept of cultural kinship

5.1 Genetic variation – meiosis, fertilisation and mutations

Variation and variability both imply the presence of genetic differences. What, then, is the difference between these two terms? In the Year 11 course, in the section on evolution, you learned that variation is evident in *individuals* (for example, differences in fur colour or height). In genetics, the term variability relates to the different forms of a gene within a *population* – that is, the total of all alleles present in the *gene pool* of a population (for example, coat colour in a population of Australian kelpie dogs includes black, red, blue or fawn, with or without tan markings)(Fig. 5.2). The genetic inheritance of coat colour in horses, rabbits and many other animals is similarly variable.


Anna Idestam-Almquist/Alamy Stock Photo

FIGURE 5.2 Australian kelpies, showing fur variability: coat colours include black and tan (considered the true colour of the working kelpie), red and black.

There are several ways in which variation arises during sexual reproduction, including during the processes of meiosis (gamete

formation) and fertilisation. Both these processes randomly mix two sets of parental chromosomes, resulting in individuals with unique and varied genotypes. Mutation is another way of introducing genetic variation, and this is discussed in Chapter 7.

Weblink
Equine genetics
Watch at least two video clips from this site about equine genetics.

Meiosis: chromosome number, variation and gamete formation

Every species has a characteristic number of chromosomes in every body cell (for example, 46 chromosomes in humans) and this number does not change from one generation to the next. **Meiosis** maintains a constant chromosome number from one generation to the next.

Meiosis is sometimes called the 'reduction division', because a diploid cell divides into four haploid daughter cells (a tetrad), reducing the chromosome number. A diploid parent cell contains two sets of chromosomes – one paternal and one maternal set. Each pair of chromosomes in the cell is termed a **homologous pair**, because the two chromosomes carry alleles for the same genes (Fig. 5.3). When homologous pairs of chromosomes align in early meiosis, each pair is called a **bivalent**.

A homologous pair of chromosomes (bivalent) consists of two similar chromosomes that carry genes for the same traits: one of the pair is maternal in origin and the other is paternal.

Worksheet
Chromosome terminology

Similarities between meiosis and mitosis

In both mitosis and meiosis:

- The names of the stages – interphase, prophase, metaphase, anaphase and telophase – are the same.
- Interphase occurs first, prior to nuclear division. During this stage, the DNA replicates, so each chromosome makes an identical copy of itself (Fig. 5.3a).
- Chromatin material transforms into chromosomes in the same way during prophase in the first meiotic division (Fig. 5.3b and c).
- The breaking down of the nuclear material and the formation of the spindle are the same.
- Cytokinesis in meiosis takes place in the same manner as in mitosis, depending on whether the cell that is dividing is a plant or an animal cell.

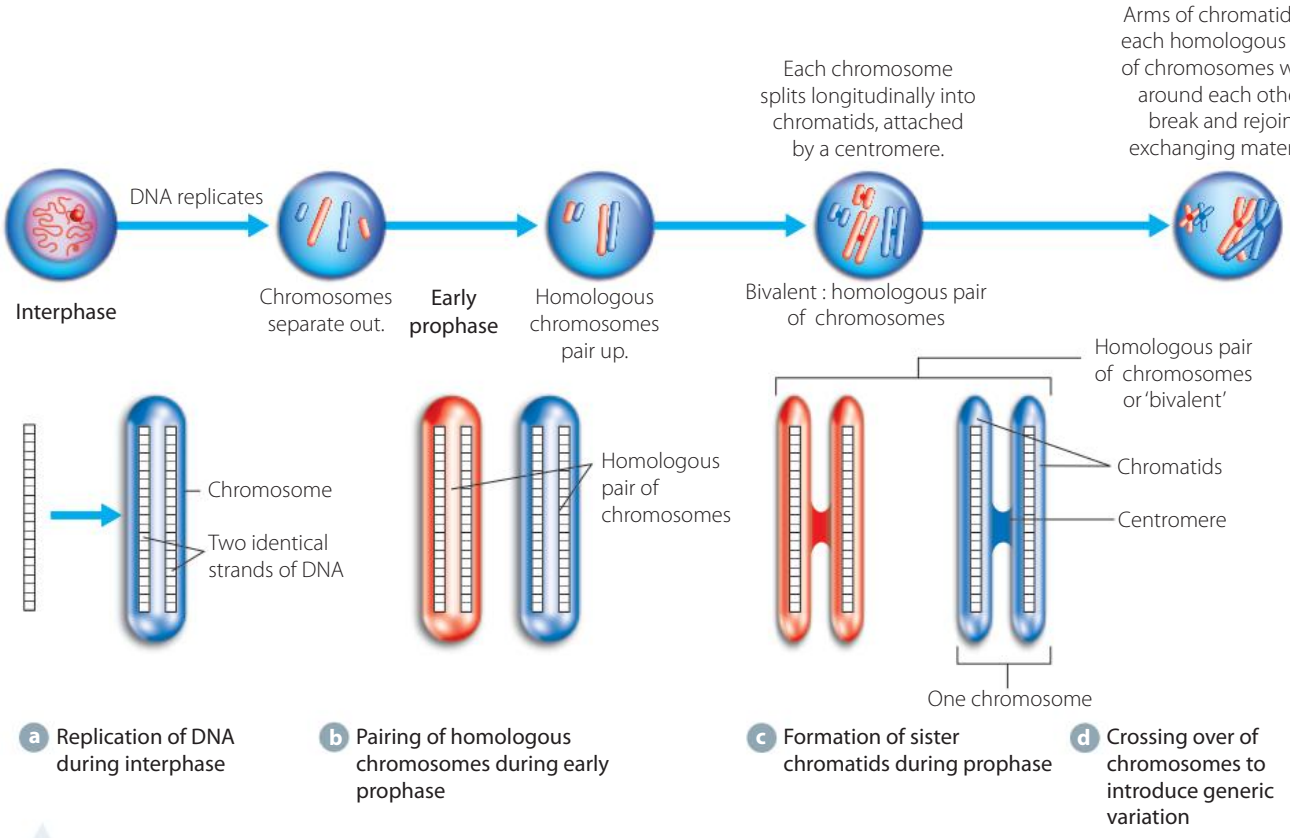

FIGURE 5.3 Early stages of division in meiosis – interphase and prophase of meiosis I

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You studied mitosis in detail in Chapter 3. Revise your understanding of terminology for mitosis using Figure 5.3 and the worksheet *Chromosome terminology*.

Worksheet
Meiosis

Meiosis I and genetic variation

Meiosis occurs in two stages: meiosis I (the first meiotic division) and meiosis II (the second meiotic division). The reduction in chromosome number occurs in meiosis I.

Meiosis I

The sequence of steps described below is summarised in diagrammatic form in Figure 5.6 (page 154).

1 Chromosomes line up in pairs (one maternal and one paternal chromosome in each pair) during prophase I.

2 **Crossing over** or **synapsis** occurs – the arms of a pair of homologous chromosomes, termed a bivalent, wrap around each other and the points at which they meet are called **chiasmata** (singular *chiasma*). The arms of homologous chromosomes break where they meet, exchanging genetic material between paternal and maternal chromosomes (Fig. 5.4).

Genes that occur on the same chromosome are said to be *linked*. Crossing over (synapsis) ensures that not all linked genes on a chromosome are inherited together. The exchange of genes during crossing over causes mixing of paternal and maternal genes and introduces genetic variation. No two chromatids are identical (Fig. 5.5).

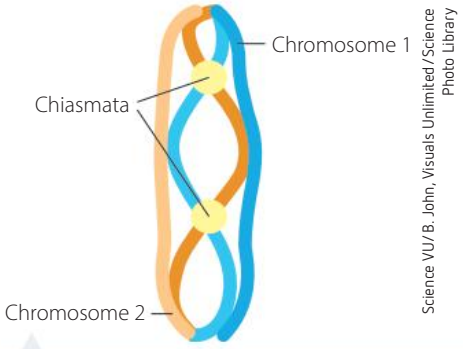


FIGURE 5.4 The points where a pair of homologous chromosomes meet when they form a bivalent during Meiosis I are called chiasmata

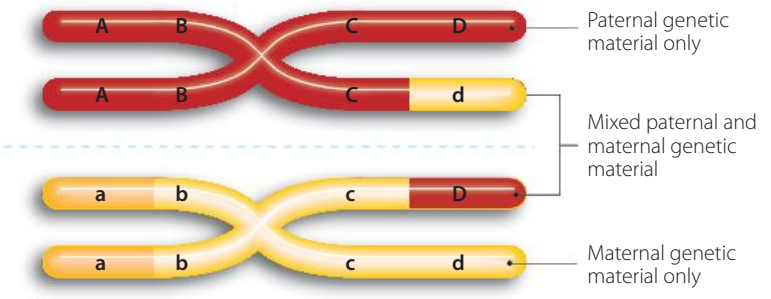


FIGURE 5.5 A bivalent (homologous pair of chromosomes) showing exchange of genetic material as a result of crossing over (synapsis), which introduces genetic variation

3 Each pair of chromosomes separates (during anaphase I), and one entire chromosome of each pair moves into a daughter cell. (Each chromosome still has two sister chromatids attached to each other.) This separation of maternal and paternal chromosomes not only halves the chromosome number in gametes, but also leads to *genetic variation*, depending on which chromosome (paternal or maternal) of each pair ends up in which daughter cell. This is termed *independent assortment* of chromosomes and produces different combinations of genes in different gametes (Fig. 5.6).

Meisois II

The two daughter cells that result from meiosis I each undergo meiosis II, which is similar to mitosis.

4 The centromere divides and the chromatids separate from each other (during anaphase), moving to opposite poles (telophase), where a nuclear membrane forms around each set of chromosomes. Cytokinesis follows, resulting in four daughter cells (a tetrad), each with half the original chromosome number. Genetic variation has also been introduced, because the combination of paternal and maternal chromatin material in each resulting daughter cell is different.

5 Many combinations of chromosomes are possible in gametes as a result of meiosis, resulting in a variety of gametes forming. Further variation is introduced during fertilisation, depending on which gametes fuse.

In the annotated diagrams in Figure 5.6, meiosis is represented in a hypothetical (model) organism that has only *two pairs* of chromosomes, keeping the representation of the process simple.

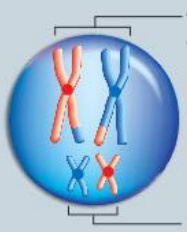
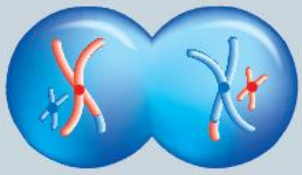
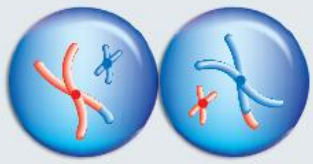
Early prophase		• Chromosomes separate into homologous pairs
Late prophase		• Nuclear membrane breaks down • Chromosomes split into chromatids • Crossing over occurs: genetic variation introduced
Metaphase I		• Chromosomes align in pairs in the middle of the cell • Random segregation • Each pair lines up independently of the next (independent assortment), so various combinations of paternal and maternal chromosome alignment are possible
Anaphase I		• Chromosome pairs separate and each chromosome moves to the opposite end of the cell
Telophase		• Two daughter cells form • Chromosome number is halved • Chromosome combinations in cells differ
Cytokinesis I		• Daughter cells are not identical to each other and have half the original number of chromosomes

FIGURE 5.6 The process of meiosis in animal cells

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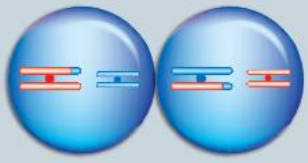
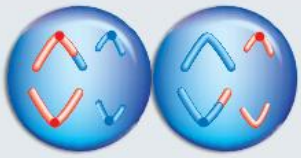
Metaphase II		• Chromosomes align at the equator
Anaphase II		• Daughter chromosomes move apart, to opposite poles
Cytokinesis II: tetrad	 Four haploid cells	• Four resulting daughter cells are not genetically identical to each other and have half the original chromosome number

FIGURE 5.6 (*Continued*)

As a result of both independent assortment and random segregation of maternal and paternal chromosomes during meiosis, further genetic variation is introduced. There are 2^{23} possible combinations of chromosomes in the formation of a human gamete, based on independent assortment only.

The process of fertilisation, which involves the random meeting of any two gametes, ensures further mixing of genetic material, producing variations in phenotype that may be acted on through natural selection in the process of evolution.

Another source of variation is mutation, which may arise at any point in the process but most commonly occurs during replication of DNA prior to the start of cell division (for example, during meiosis).

Fertilisation and new combinations of genotypes

Gametes are haploid and contain different, recombined genetic material in their single set of chromosomes. This variation is introduced by synapsis and independent assortment during meiosis. In the process of fertilisation, there are many possible combinations of gametes that may fuse (depending on which sperm cell fuses with which egg cell). This further increases variation within the individual in the form of new combinations of genes, leading to greater *variability* within the population.

Self-fertilisation or cross-fertilisation?

Sexual reproduction between genetically dissimilar parents of the same species produces offspring that are likely to differ from each other more than offspring produced by sexual reproduction where male and female organs are on the same individual. For example, offspring arising from cross-fertilisation between plants will have greater genetic diversity than those that arise from self-fertilisation, and offspring arising from gametes produced by unisexual animals will have greater genetic diversity than those arising from hermaphroditic (bisexual) animals.

INVESTIGATION 5.1

Information and communication technology capability

Literacy

Weblink
Independent assortment and gamete diversity
Watch the animation, listen to the narration and then work through the quiz.

Weblink
Meiosis with crossing over

Secondary-source and practical investigation to model meiosis, fertilisation and mutations

During meiosis, genetic variation arises as a result of the behaviour of chromosomes during:

- synapsis and crossing over
- independent assortment and random segregation.

In your investigation, you need to model how variation is introduced in the process of meiosis, fertilisation and mutation.

AIMS

1. To model meiosis (including crossing over, independent assortment and random segregation) and predict variations in the gametes produced (Part A)
2. To model fertilisation in order to predict variations in the genotype of offspring (Part B)
3. To predict variations in the genotype of offspring if a mutation was to arise during meiosis and/or subsequent fertilisation (Part C)

RESOURCES

To research how meiosis may be modelled and gain ideas for creative approaches, refer to secondary source material. This may include information in this textbook, as well as online websites, video clips and animations, such as the weblink resources.

PART A: CHROMOSOME BEHAVIOUR DURING MEIOSIS LEADS TO GENETIC VARIATION

AIM

To model meiosis, including crossing over, segregation, independent assortment of chromosomes and the production of haploid gametes

MATERIALS

Pipe cleaners (or playdough/plasticine/strips of paper) in two different colours, to represent chromosomes – one colour represents paternal and the other maternal chromosomes. Make each homologous pair of chromosomes a different length – long, medium and short. To keep your model simple, demonstrate meiosis in a parent cell with *three* pairs of chromosomes.

METHOD

1. In your model of meiosis, use a cell with:
   - at least three pairs of chromosomes made out of pipe cleaners, strips of paper or strings of beads
   - two colours to distinguish between maternal and paternal chromosomes
   - different-sized chromosomes to distinguish between the three homologous pairs (for example: long, medium and short).
2. Move the chromosomes through each stage of meiosis on templates of cells drawn on A3 paper, or use the Modelling meiosis template worksheet. Select a method of recording your results from ideas suggested in the results section.

Model how variation is introduced by the process of meiosis, showing the processes described below.

Meiosis I

1. *Crossing over* – linked genes are exchanged between paternal and maternal chromosomes, increasing the possible combinations of genotypes.

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2 *Independent assortment* – chromosomes line up across the cell centre in homologous pairs, with paternal and maternal assortment being independent.

3 *Random segregation* and halving of the chromosome number – whole chromosomes in each bivalent separate and move into different cells, with random separation of paternal and maternal chromosomes.

There are 2^{23} possible combinations of chromosomes in the formation of a human gamete. Predict how many combinations would be possible in your hypothetical cell with three pairs of chromosomes.

Meiosis II

The two daughter cells that result from meiosis I each undergo meiosis II, which is similar to mitosis, but each cell ends up with its own unique set of daughter chromosomes.

Whether annotating or narrating your model, make sure you use the correct scientific terminology in context. You may wish to make a list of the terminology that you will use to describe each stage of meiosis, referring to the glossary of terms at the back of this textbook to assist you before you begin annotating or narrating.

RESULTS

Record your model using one of the options below.

Option 1: Draw around each chromosome, using coloured pencils, at each stage of your model. Label all diagrams and provide a heading for each. Write a brief annotation outlining what is happening in each stage.

Option 2: Use a digital camera or your mobile phone to take a photograph of each stage of meiosis as you model the process. Print photos and arrange them in a table (use Table 5.1 as a guide). Label each diagram and provide a brief annotation outlining what is taking place at each stage.

Option 3: Use a digital camera or your mobile phone to make a video of the process of meiosis as you model it. Create a voice over to narrate what is happening in your video.

Option 4: Use a stop motion software app on an iPad or iPhone to record your model, and narrate the presentation or add your own text. (You may wish to view YouTube clips to learn how to use the app.)

Worksheet
Modelling meiosis: template

TABLE 5.1 Results of modelling meiosis

STAGE OF MEIOSIS	DIAGRAM/PHOTOGRAPH	ANNOTATION

COMMUNICATING YOUR INVESTIGATION

Write a practical report, outlining the aim, materials, method, results and conclusion. In your report, include the answers to the discussion questions below.

DISCUSSION

1 Explain why two different colours were used for chromosomes.

2 Draw a diagram of the stage where the chromosome number was halved and describe how this was modelled.

3 Draw alternative arrangements of these chromosomes as they undergo independent assortment and segregation, to show all possible variations of gametes that may arise.

4 Predict how many combinations would be possible in your hypothetical cell with three pairs of chromosomes. Explain your reasoning.

5 How many chiasmata are visible in the bivalent in Figure 5.6? Draw a diagram to predict the result of this crossing over.

6 Assess the validity, accuracy and limitations of your model.

Review accuracy, precision and errors in measurement in Chapter 1 page 16.

PART B: FERTILISATION INTRODUCES GENETIC VARIABILITY

1 Design and develop a model to demonstrate how fertilisation may introduce genetic variability in offspring. You may wish to use the gametes you created in your meiosis model to demonstrate how fertilisation introduces genetic variation. Predict the possible variations that may arise in the genotypes of offspring.

2 Test your prediction and record the results.

3 Explain any limitations in data obtained.

4 Evaluate how you could improve your investigation plan.

PART C: PREDICTING VARIATIONS CAUSED BY MUTATION

1 Develop a model to demonstrate mutation during meiosis and predict the variation in the genotype of offspring. (You may wish to make adjustments to the design of your model, taking into account your answer to Question 4 in Part B.)

2 Test your prediction and record the results.

3 Evaluate the benefits and limitations of your model.

KEY CONCEPTS

- The genetic consequences of meiosis:
  - One cell undergoes two meiotic divisions to generate four haploid cells.
  - The genes in each haploid cell are a new combination of the parental genes.
  - The new combination results from both crossing over and random segregation, allowing the individual alleles of maternally and paternally derived chromosomes to assort independently.
- The genetic consequences of fertilisation:
  - Two haploid gametes fuse to form a diploid zygote.
  - The genes in the zygote are a combination of the genes contributed by the parents: 50% paternal and 50% maternal (not taking into account material that was exchanged during crossing over).
- Variability:
  - Variations in gene content of the gametes give rise to individuals in the population with new gene variations, increasing variability.
  - Mutations may further contribute to genetic variation in an individual and genetic variability within a population.
- Models of meiosis are simplifications of the actual process, designed to demonstrate specific aspects, such as the introduction of genetic variation. These models have limitations, for example in not demonstrating all aspects of a process.

CHECK YOUR UNDERSTANDING 5.1

1 Draw a Venn diagram to compare the processes of mitosis and meiosis. Suggested areas to consider are the:

- type of cells in which division occurs
- number of divisions and resulting daughter cells
- chromosome behaviour:
  - prior to division (interphase)
  - in the early stages of division (prophase)
  - during segregation (metaphase/anaphase of the first meiotic division)
  - at the end of cytokinesis (mitosis) and cytokinesis I and II (meiosis)
- end result – number of cells; number of chromosomes.

2 Explain the biological importance of crossing over during meiosis.

3 Describe how two processes other than meiosis in sexual reproduction introduce genetic variation.

4 Calculate how many combinations of chromosomes would be possible in the gametes formed when a cell with eight chromosomes undergoes meiosis. Show your working.

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5.2 Genotypes and inheritance patterns

Inheritance patterns have been a subject of fascination for many years. Mendelian inheritance, proposed over 170 years ago by Gregor Mendel, is the basis of all inheritance patterns. Using mathematical calculations, Mendel proposed a model whereby one could *predict the ratios* of various types of offspring from any two specific parents.

When Charles Darwin and Alfred Russel Wallace proposed their theory of evolution by natural selection in 1858, there was no knowledge of what was responsible for the differences in individuals within a population or of how such variations in characteristics could be passed from one generation to the next.

Once Mendel's laws were explained in terms of modern genetics, the model known as **modern synthesis** arose, combining the understanding of Mendelian genetics with Darwinian evolution and giving us our modern-day theory of evolution (sometimes called neo-Darwinism).

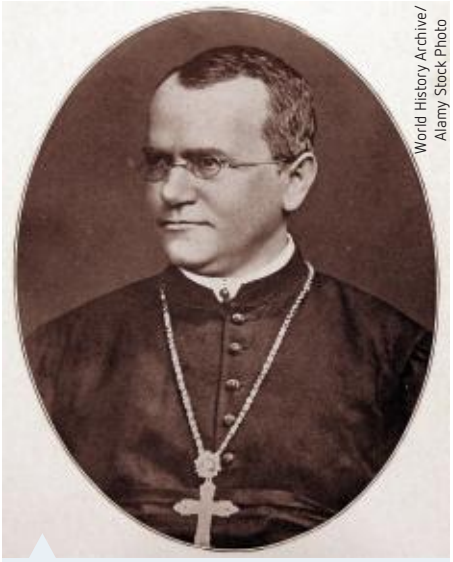
World History Archive/ Alamy Stock Photo

FIGURE 5.7 Gregor Mendel, the father of genetics

Autosomal recessive inheritance

Mendel's model of inheritance was based on a specific set of conditions and this model still holds true today if these same conditions prevail – this pattern of inheritance is known as **autosomal recessive inheritance**.

Autosomal recessive inheritance occurs under the following conditions:

- A version of each characteristic or trait in an individual is inherited from both parents and is therefore controlled by a pair of inherited *factors* (called **alleles**).
- Alleles pass from one generation to the next according to set ratios.
- The alleles in an individual may be the same (in pure-breeding or **homozygous** individuals) or may differ (in hybrid or **heterozygous** individuals).
- In hybrid individuals, the trait that is *expressed* (appears) is known as the **dominant** allele, whereas the one that is hidden or *masked* is the **recessive** allele (Mendel's first law – dominance). For a recessive trait to be expressed, both alleles in an individual need to be recessive.
- During gamete formation, the pair of alleles for a trait segregate (separate) and each gamete receives only one allele for the trait/gene (Mendel's first law – segregation).
- When the inheritance of more than one trait/gene is studied, the pairs of alleles for each trait separate independently of the other pairs of alleles (Mendel's second law – independent assortment).

Worksheet
Mendel's laws

Modern genetics terminology used to describe inheritance patterns

In modern-day studies of genetics, we know that different genes influence different characteristics. For example, in pea plants, one gene may determine seed colour, while another determines stem length. In humans, a range of different alleles for each gene may determine characteristics such as height, eye colour and hair colour.

- *Genes* on chromosomes determine characteristics that are inherited.
- *Alleles* are different forms of the same gene and occur in pairs in diploid individuals.
- Alleles are found in identical positions or **loci** (singular **locus**) on pairs of homologous chromosomes within cells (Fig. 5.8).
- *Diploid* individuals have two alleles of each gene, and *haploid* cells (gametes) have only one allele of each gene.
- The **phenotype** of an organism, simply put, is its appearance (the expression of an organism's genes). The **genotype** of an organism is the combination of genes that is present in each cell.

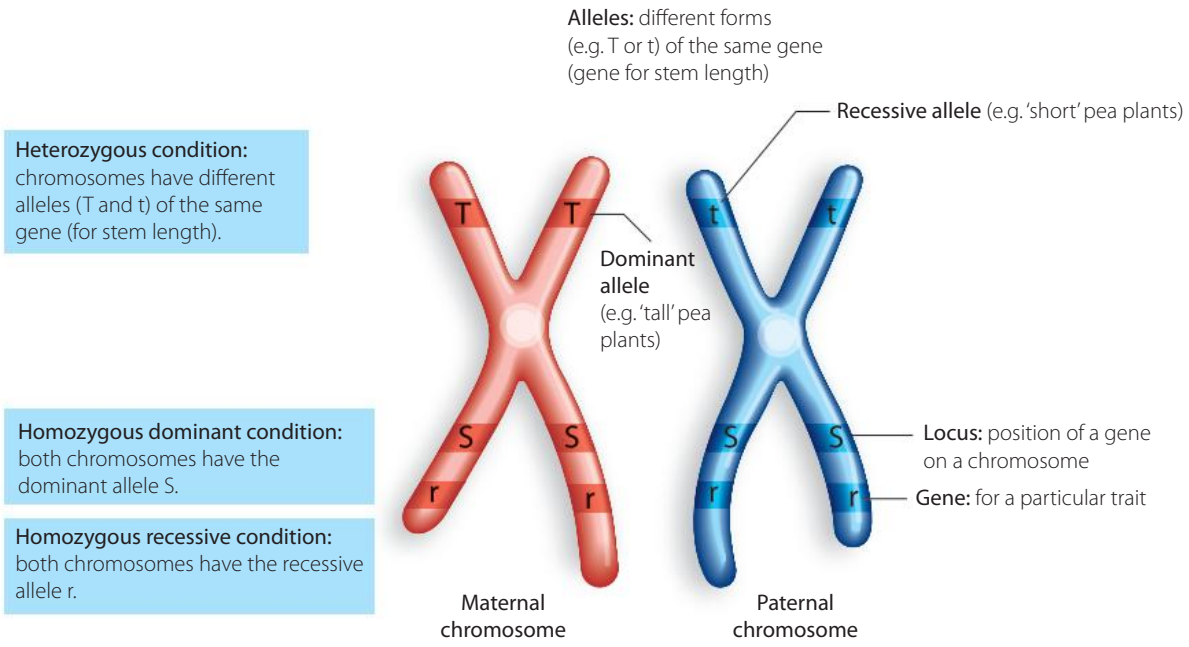

FIGURE 5.8 Pairs of chromosomes showing alleles, gene, locus, and homozygous and heterozygous genotypes

Today we know that, at a molecular level, the *phenotype* is more complex than just an organism's appearance. It is the sum of the gene products (proteins and RNA) that are made and these give rise to not only the physical appearance, but also the behaviour and functioning of organisms.

The phenotype of an organism is determined to a large extent by the genetic makeup or *genotype* of the individual, but the phenotype may also be influenced or modified by interaction with the *environment*. For example, the final height of a human adult depends on a combination of that person's *genotype* as well as their *nutrition* – if they are underfed or receive insufficient protein while growing, they may never attain their full potential height as determined by their genotype. (See Chapter 4.)

Autosomal recessive inheritance and genetic crosses

Autosomal recessive inheritance was proposed as a result of Mendel's experiments with garden pea plants. He investigated their breeding patterns to determine the inheritance of a variety of characteristics or traits. Pea plants are ideally suited because they can easily be grown and cross-bred, they have a short life cycle (they are annual plants), and their flowers have both male and female parts.

Mendel studied the inheritance of each trait individually (for example, trait = stem length), investigating the inheritance of one pair of contrasting features at a time (for example, tall or short stem length). He studied the following traits and their alternative forms:

- stem length – tall or short
- colour of seed contents – yellow or green
- colour of seed coat – grey or white
- shape of seeds – round or wrinkled peas
- colour of unripe pod – yellow or green
- flower position – axial or terminal
- pod shape – inflated or constricted.

The traits Mendel studied were typical examples of the autosomal recessive inheritance pattern we know today. That is, if two alleles of a gene are present in a population, one allele is *dominant* (seen in

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the phenotype) and the other allele is masked or *recessive*. Autosomal recessive inheritance also assumes that these alleles are located on one of the *non-sex chromosomes* (autosomes).

For simplicity, a letter of the alphabet is used to represent each allele of a trait (such as stem length in plants). The different alleles are distinguished by using a capital letter for the dominant allele (for example, *T*) and a lower case version of the *same letter* for the recessive allele (*t*).

Note: Capital and lower case versions of the same letter signify dominant and recessive alleles of the same genetic trait.

Mendel's monohybrid cross

Mendel carried out a pure-breeding cross followed by a monohybrid cross (Fig. 5.9). That is, he crossed two parents (P) who were **pure-breeding** (homozygous) for both characteristics – for example, TT and tt. All offspring (F_1 or first filial generation) appeared phenotypically tall, but when they were cross-bred, their offspring (F_2 or second filial generation) gave the phenotypic ratio of 3 tall:1 short.

Mendel used mathematical calculations to show that this type of inheritance pattern required one factor to be passed on from each parent to the F_1 generation, who were hybrids (Tt). When these F_1 hybrids were bred, they gave a genotypic ratio in the F_2 generation of 1TT:2Tt:1tt (Fig. 5.9). It was from these ratios that Mendel derived his first law of dominance and segregation.

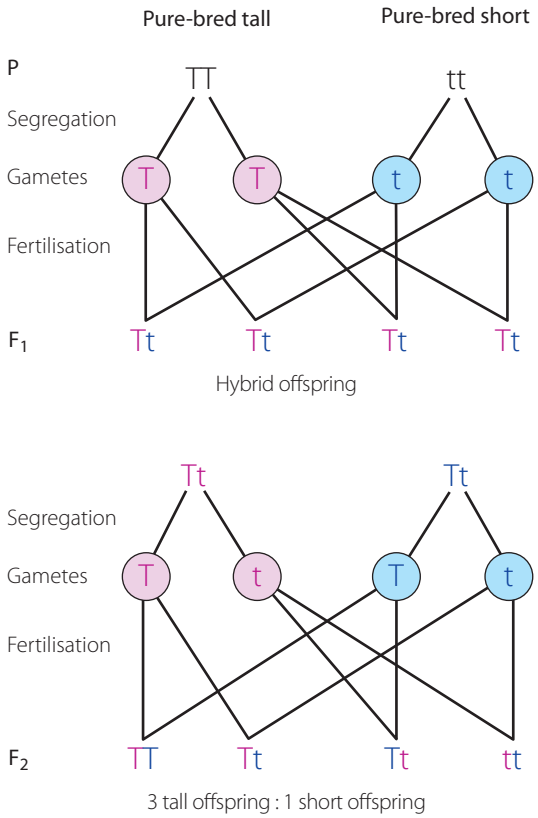

FIGURE 5.9 Mendel's monohybrid cross showing autosomal recessive inheritance

Mendel's laws

What makes Mendel's discoveries so remarkable is that chromosomes and genes were not discovered for another 35 years. His laws still hold true today. Theories and laws in science are concepts founded on clearly identified assumptions, can be tested experimentally and give reproducible results. They can be used to explain and predict a wide range of observed phenomena.

Refer to Chapter 1 page 5, 'Models in science'.

Mendel's first law of dominance and segregation

The characteristics of an organism are determined by factors that occur in pairs. Only one member of a pair of factors can be represented in any gamete (*segregation*). Offspring inherit one factor from each parent. When two hybrids breed, statistically they will produce a ratio of three offspring showing the same trait as the parents (termed the *dominant* trait) to one offspring showing the contrasting *recessive* trait – that is, the ratio of dominant to recessive trait will be 3:1.

Mendel's second law of independent assortment

Mendel's second law relates to how variation arises in meiosis. Mendel carried out more complex experiments involving *dihybrid* crosses to establish his second law, studying two pairs of factors and their separation. In this law, he established further ratios showing that when individuals with *two* or more pairs of unrelated, contrasting characteristics are crossed (for example, tall plants with yellow pods × short plants with green pods), the different pairs of factors (tall/short and yellow/green) separate out independently of each other. That is, tall is not always inherited with yellow and short with green – some tall green offspring and some short yellow offspring will result. This law assumes that the genes are located on different chromosomes.

Autosomes and sex chromosomes

Every cell in the human body contains 23 pairs of chromosomes: 22 pairs of autosomes (chromosomes that code for general traits within the body) and 1 pair of **sex chromosomes** (Fig. 5.10). Sex chromosomes carry genes that determine the sexual characteristics of a person and therefore influence whether they are biologically male or female.

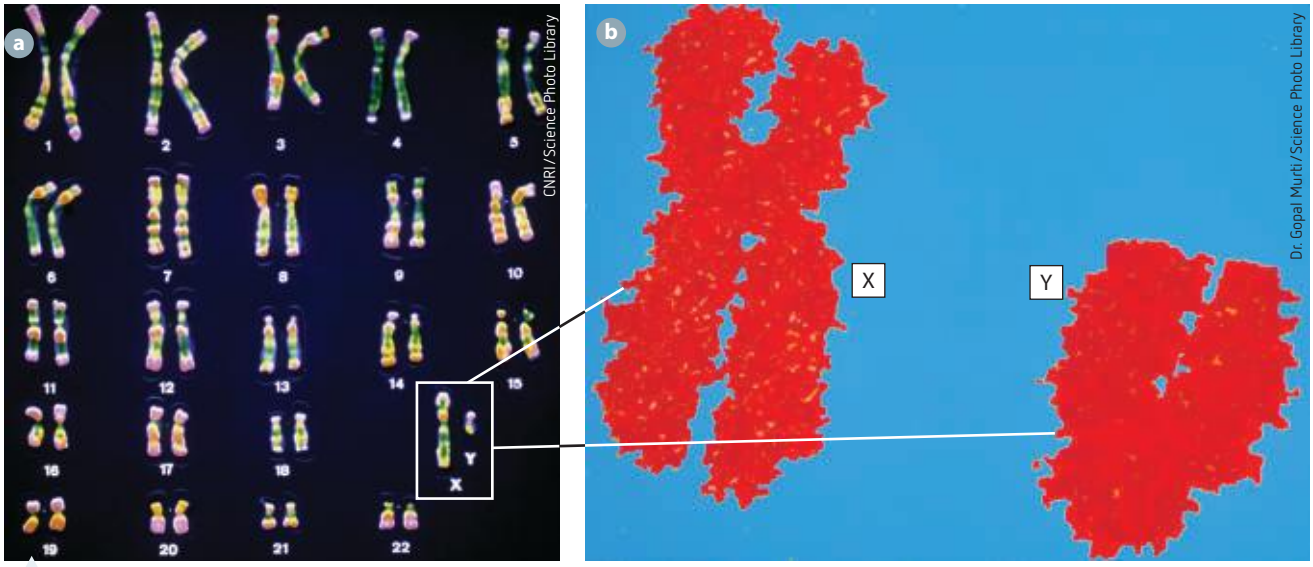

FIGURE 5.10 **a** The human karyotype of a male; **b** scanning electron micrograph image of human X and Y chromosomes

Sometimes within a *population* there are more than two alleles for a particular gene. For example:

- alleles for flower colour in sweet peas – pink, white, purple, red
- alleles for hair colour in Labrador dogs – black, brown or yellow (golden).

However, an *individual* can have only two alleles, and which alleles they possess depends on which pair of alleles have been passed on to them by their parents. Multiple alleles in a population give the group greater genetic variability and result in greater diversity.

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KEY CONCEPTS

- A gene:
  - is a segment of DNA on a chromosome
  - specifies a particular characteristic (e.g. seed colour)
  - has two alleles in an individual and two or more alternative alleles in a population.
- Alleles:
  - are alternative forms of the same gene
  - occur in pairs in a diploid individual, but two or more alleles for each gene may be present in a population
  - segregate during gamete formation (meiosis)
  - occur individually in each haploid gamete
  - pair during fertilisation, when the diploid condition of an organism is restored during zygote formation.
- Homozygous and heterozygous genotypes:
  - Mendel's terms 'pure-breeding' and 'hybrid' are known in modern genetics as 'homozygous' (e.g. TT or tt) and 'heterozygous' (e.g. Tt) respectively.
  - The term 'homozygous' is derived from two words: *homo* = the same; and *zygote* = a fertilised egg that has received half its genetic material from each parent.
- Genotype and phenotype:
  - The genetic makeup or genotype of an organism determines its physical appearance or phenotype.
  - Phenotype is not only the physical appearance of an organism, but may also include its physiology (functioning) and aspects of its behaviour. On a molecular level, phenotype is the sum of the gene products (proteins and RNA) that are made.

CHECK YOUR UNDERSTANDING 5.2a

1. How did the model known as modern synthesis arise?
2. Explain autosomal recessive inheritance, using an example.
3. Allocate letters to represent each of the alleles of the characteristics that Mendel studied in pea plants. Assume the first allele is the dominant allele.
4. In poodles, black coat colour is dominant to white coat colour. Assign letters to represent each allele. Following Mendel's laws and the genetic crosses shown in Fig 5.9, work out what the offspring would look like if you crossed a homozygous black poodle with a white poodle. What genotypic ratio would you expect?
5. Distinguish between autosomes and sex chromosomes.

Solving genetics problems

Genetic problems come in many forms. To solve a genetics problem, you need to use the information given – read it, analyse it and see what deductions you can make from it. To make predictions in any genetic cross, it is important to determine which trait is dominant – this information may be known (visible in the phenotype), or it may be worked out by deductive reasoning. Sometimes it remains unknown.

Representing autosomal inheritance using a Punnett square

A useful problem-solving technique for organising information and representing a genetic cross is to draw a Punnett square. (See Worked example 5.1, on page 164.)

A Punnett square is a model used to represent inheritance patterns such as autosomal inheritance, as shown in Mendel's monohybrid cross. Models such as this can be used in predicting possible outcomes when certain individuals are cross-bred. This is a very useful problem-solving tool, as it allows scientists to calculate the probability of a genetic trait (such as blue eyes or dimples) being passed on.

Punnett squares are often used to calculate the probability that a genetic defect will be inherited by offspring of two parents of known genotype. A Punnett square shows the basic pattern of inheritance – how

two factors in each parent separate during the formation of gametes (segregation) and the possible combinations of alleles that could arise during fertilisation. The probability of any particular genotype or phenotype arising can then be calculated from the results.

Probability in genetics

You have seen how the phenotypic ratio of offspring (3 dominant:1 recessive) was derived from Mendel's typical monohybrid cross. This means that, taking the gene for height as an example, three-quarters of the offspring will be tall and one-quarter will be short. The probability can also be written as a percentage – there is a 75% probability that offspring will be tall and a 25% probability that they will be short.

This result will only be obtained with a large sample size, as it is based on probability. Even if a large number of plants are studied, the ratio will be approximate. For example, when Mendel studied height in garden peas, he obtained 787 tall and 277 short plants (a ratio of 2.84:1). For yellow peas and green peas, he obtained 6022 yellow peas and 2001 green peas (3.01:1). These ratios are close to, but not precisely, 3:1.

The more individuals that are counted, the more closely the observed ratios approach the expected values.

Mendel's ideas and logic can easily be represented using a Punnett square to predict the features of offspring in a genetic cross, as shown in the worked example.

WORKED EXAMPLE 5.1

1 Use a Punnett square to predict the ratio of offspring produced if two individuals that are heterozygous for height are crossed.

ANSWER	LOGIC
Let T = tall and t = short F_1: Hybrid tall × Hybrid tall Tt Tt Segregation Gametes T t T t	▪ Write the parent genotypes that are to be crossed. ▪ Show how the gametes segregate during meiosis.
Gametes: T, t T: TT, Tt t: Tt, tt TT : Tt : Tt : tt 3 tall : 1 short	▪ Draw up a 3 × 3 grid. Write the genotypes of one set of gametes across the top of the grid and the other set of gametes down the side. ▪ In the centre of the grid, write the genotypes that would result from each combination of gametes. ▪ Write the genotypic ratio. ▪ Write the phenotypic ratio.

TRY THESE YOURSELF

1 In Labrador dogs, black hair, B, is dominant to white hair, b. Predict the expected genotypic and phenotypic ratios resulting from the crosses below.

a homozygous black dog × white dog

b two heterozygous black dogs

c heterozygous black dog × white dog

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2 In fruit flies, curly wing is dominant to straight wing. Two curly-wing fruit flies produce 51 offspring with straight wings and 149 curly-wing offspring.

- **a** What are the probable genotypes of the parent flies? (Hint: Work out the ratio of curly-wing to straight-wing flies.)
- **b** Justify your answer using a Punnett square.
- **c** Calculate what proportion of the curly-wing offspring would be heterozygous.

Worksheet
Dominant and recessive traits

A test cross

When you consider an organism's phenotype, it is not always possible to tell what its genotype is. For example, in Mendel's pea plants, a phenotypically tall plant may have the genotype TT or Tt. To determine the genotype in cases such as this, geneticists use a technique called a **test cross**.

For example, in the fruit fly *Drosophila melanogaster*, wing length is determined by a single gene. The allele for long (normal) wings (Fig. 5.12a) is dominant and can be represented by the letter V. The allele for short (vestigial) wings (Fig. 5.12b) is recessive and can be represented by the letter v. To determine whether a fly with long wings is homozygous (VV) or heterozygous (Vv), it can be crossed with a fly that is homozygous for the recessive gene (vv). Using Mendel's expected ratios, we can predict that if the fly is homozygous for long wings, all offspring will have long wings, because they will all inherit a V allele from the long-winged parent and a v allele from the short-winged parent, resulting in all offspring having the genotype Vv.

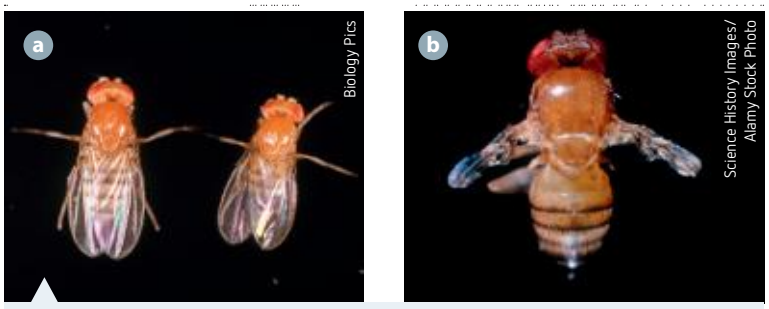

FIGURE 5.12 *Drosophila* fruit flies with **a** normal wings and **b** vestigial wings

However, if the long-winged parent fly is heterozygous (Vv), it will pass one V allele to half its offspring and one v allele to the other half. The offspring will all receive a v allele from the short-winged parent. Therefore the ratio of long-winged (VV or Vv) to short-winged (vv) offspring will be 1 : 1.

The two crosses can be represented in a Punnett square, as shown below.

Weblink
Probability of inheritance
Do the quiz, working through the questions and Punnett squares.

Cross 1

P: VV × vv

Gametes	V	V
v	Vv	Vv
v	Vv	Vv

100% of F_1 offspring are Vv and therefore have long wings.

Cross 2

P: Vv × vv

Gametes	V	v
v	Vv	vv
v	Vv	vv

50% of F_1 offspring are Vv and have long wings, 50% of F_1 offspring are vv and have short wings.

Test crosses are carried out between an organism that exhibits a dominant trait and an organism that exhibits a recessive trait, to determine whether the dominant organism is homozygous or heterozygous for the dominant trait.

Pedigree analysis

If the traits expressed in a family over several generations are observed, a **pedigree chart** (family tree) can be constructed to record phenotypes. This may be used to work out the genotypes of family members, some of which may be more easily determined and some of which may be unknown. A pedigree chart and analysis is often used to study heredity patterns in families by tracing the inheritance of any particular characteristic, and to make predictions about the expected phenotypes and genotypes of future offspring.

Pedigree charts can be used to identify inheritance patterns in a variety of organisms. Their usefulness may range from studying mutations in humans that result in disease or disorders, to studying desirable phenotypic traits in horses for breeding purposes. The traits that appear in each generation are recorded using particular symbols and lines to show relationships. Analysis of pedigree charts is based on using logic and reasoning to work out the genotypes of parents and offspring.

It is often necessary to study at least three generations to find out the genotypes of individuals. This is because masked recessive genes cannot be detected by simply examining the phenotype of one or two individuals. Studying a trait over several generations may reveal the genotypes, because recessive traits may be masked and appear to 'skip' a generation, reappearing in later generations. (See Fig. 5.14 and Worked example 5.2.)

Pedigree charts or diagrams are drawn in a universally accepted scientific format, using standard symbols. They show an individual's biological relatives and their partners as a series of circles and squares, linked by lines. The occurrence of a particular trait within the family is represented by shading.

A pedigree can therefore be defined as a graphical representation of the inheritance patterns of a particular trait in related individuals over a number of generations. Analysis of the pedigree chart is carried out to record:

- how many family members have the trait
- the gender (male or female) of the affected individuals
- how individuals in the pedigree are related.

This information may be used to:

- determine inheritance patterns
- assign genotypes to individuals where possible
- make predictions about the probability (sometimes the risk) of an individual inheriting a trait (for example, a genetic disorder, abnormality or disease).

Constructing pedigrees and analysing inheritance

Constructing and analysing a pedigree chart (Fig. 5.13) for a particular trait is done through a series of steps.

1. Gather phenotypic records of that trait in family members over several generations.
2. Use symbols to represent the various family members, showing whether they are male (square) or female (circle) and whether or not they possess the trait being studied (shaded = trait being studied is present).
3. Assign a number (Roman numeral) to each generation of the family tree and another number (Arabic numeral) to each individual in that generation.
4. Use linking lines to represent relationships between people – marriage (horizontal line) and offspring (vertical line).

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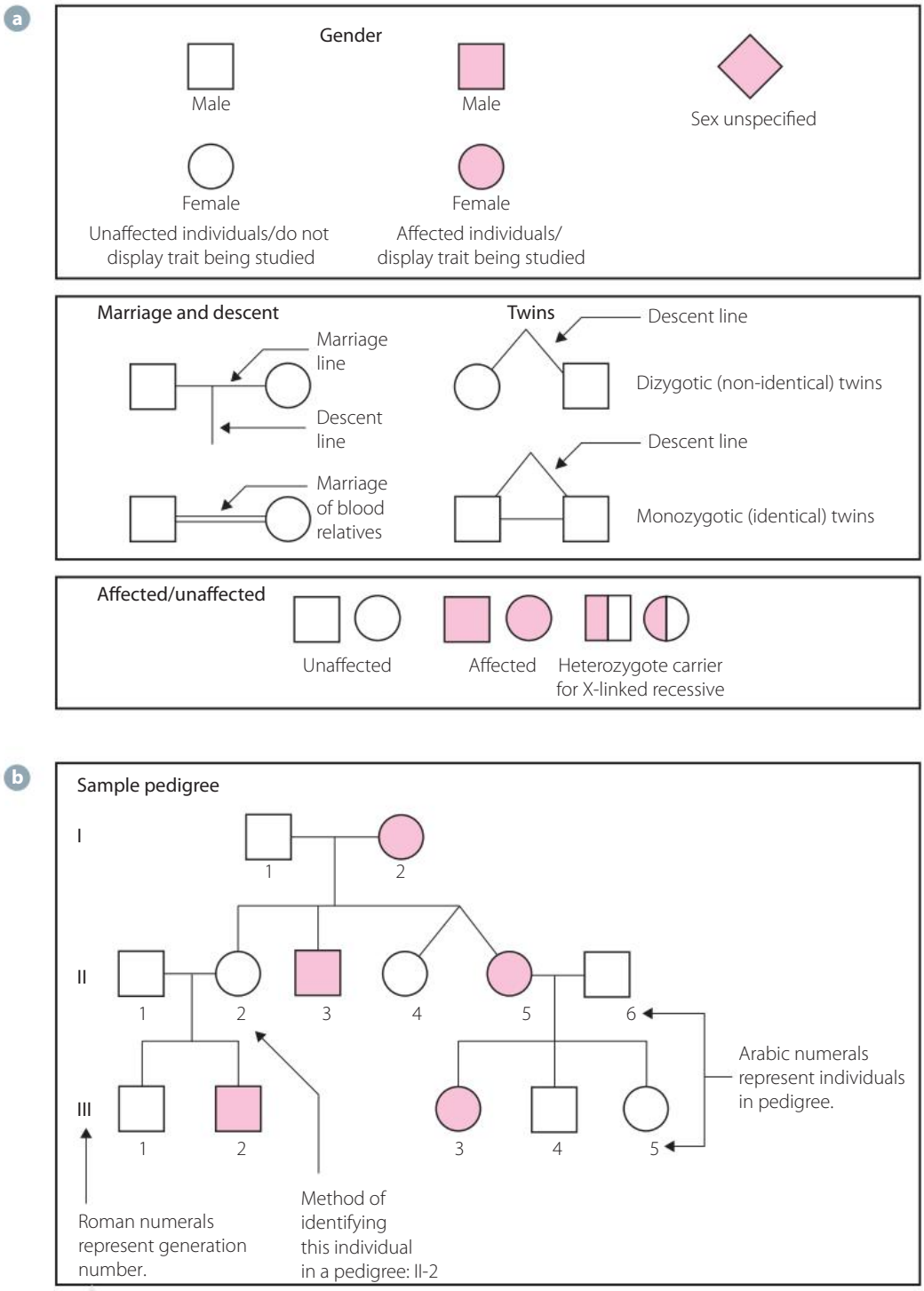


FIGURE 5.13 **a** Symbols used in human pedigree construction and analysis; **b** a sample pedigree

Table 5.2 summarises features of autosomal inheritance patterns for dominant and recessive genes. Use this table to assist with logical deductions that you need to make when analysing pedigree charts.

TABLE 5.2 Some inheritance patterns evident from pedigree analysis

TYPE OF INHERITANCE	FEATURES	EXAMPLES
Autosomal	Trait follows Mendelian inheritance pattern.	
Autosomal recessive	• The trait is expressed as the result of a recessive gene. • The affected individual must carry *two affected alleles* (be homozygous recessive) for this gene to be expressed. • The gene can be passed on to *both males and females* in equal proportions. • The trait appears to *skip a generation*.	Albinism Tay-Sachs disease Cystic fibrosis Sickle cell anaemia
Autosomal dominant	• The trait is expressed as a result of a dominant gene. • The affected individual must carry *at least one affected allele* for this gene to be expressed. • The gene can be passed on to *both males and females* in equal proportions. • The trait does not *skip a generation*.	Huntington's disease Achondroplastic dwarfism Polydactyly

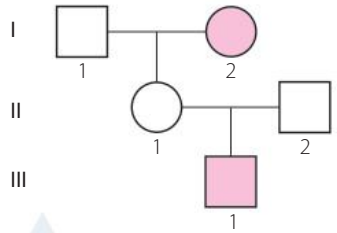
WORKED EXAMPLE 5.2

Figure 5.14 shows three generations of one family. If the trait of fair hair colour is selected for study:

i Construct a pedigree for the family shown.

ii Analyse the pedigree to determine the genotype with respect to hair colour (where possible) of each individual in the pedigree chart.

FIGURE 5.14 Family photo showing how hair colour may 'skip' a generation . The daughter in the middle of the photo is with her parents on the left and her husband and child on the right.

ANSWER	LOGIC
i Pedigree  **FIGURE 5.15** Family pedigree showing hair colour	To construct a pedigree chart: ▪ Record the phenotype of each individual – males are represented by a square, females by a circle. ▪ Shade individuals who express the trait being studied – in this case, fair hair. ▪ Use linking lines to represent relationships between people. ▪ Assign a Roman numeral to each generation of the family tree and a number (Arabic numeral) to each individual.

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ii Genetic cross and analysis *Cross from phenotype* Let B = black hair and b = fair hair P: B_ × bb (grandparents) F_1: B_ × B_ (parents) F_2: bb (grandson) *Analysis* Two individuals that express the dominant trait produce an offspring that expresses the recessive trait. *Deduction* The parent individuals with the dominant trait must have been heterozygous for that trait (each passed a 'b' allele to their son). *Genotypes* P: Bb × bb F_1: Bb × Bb F_2: bb	To analyse the pedigree chart and determine the genotypes of individuals: ▪ Record the known genotypes, working these out from the phenotypes given. Let B represent dark hair and b represent fair hair. If an allele is unknown, put an _ (underscore). ▪ Analyse the cross, using logic. ▪ Make a deduction (conclusion), using reasoning. ▪ Complete the cross by filling in the missing genotypes (red alleles) that you determined.

TRY THESE YOURSELF

1 Skippy the kangaroo is descended from a line of kangaroos that have a history of albinism. Albinos are unable to make pigment and therefore their coat colour is white. Skippy has brown fur and he mates with a brown female kangaroo named Bouncer. They produce two offspring: joey 1 is an albino and joey 2 has brown fur. Skippy's mother was an albino and his father had brown fur. Both of Bouncer's parents had brown fur.

a Construct a family pedigree to show the inheritance of albinism in the kangaroo family described.

b Analyse the pedigree to determine the genotype (where possible) of each individual in the pedigree chart. Explain your reasoning.

Worksheet
Interpreting pedigrees

INVESTIGATION 5.2

Practical investigation to gather data and construct a family pedigree chart

Worksheet
Traits and information that may be used for constructing pedigrees

Pedigree charts are used in science to trace the appearance of a phenotypic trait across several generations, to determine the type of inheritance pattern of the trait. You are to represent an autosomal trait across a family of three generations and include a total of at least seven individuals.

AIM

To construct a pedigree or family tree to trace the inheritance of selected characteristics

METHOD

1 Select a genetically determined trait in a family to construct a family pedigree chart. Conduct research to decide on a suitable genetic trait to use for gathering data for this purpose. Traits that are coded by a single gene and that have two allelic variations tend to be easiest to study. Use your own family or the family of a friend or neighbour. You will need to interview them to gather information about other family members.

»

» 2 Using correct scientific notation, construct a family tree to show the inheritance of the characteristic you have selected. If you do not have direct data about some family members, predict their characteristics using any information you have (such as photos, stories, your own traits). Use the correct symbols to represent individuals, the correct lines to show relationships and the correct numbering system for generations and individuals.

Important note: The pedigree chart must contain at least three generations and at least seven individuals.

3 Carefully label your pedigree chart with the names of family members (first names only, for confidentiality, or change the names). Also include an illustration of the characteristic you selected, such as a diagram or photograph and/or a brief description.

RESULTS

4 Analyse your pedigree chart to determine whether the gene responsible is dominant or recessive, or if dominance cannot be determined, explain why.

5 Using information in your pedigree chart, write:

- **a** three questions that other students could answer, based on the pedigree you have constructed
- **b** two questions that they would need more information to answer
- **c** one question that could form the basis of further research.

As a start, you may wish to consider whether the trait is dominant or recessive, which genotypes of family members are known and unknown, the scientific basis for the phenotype, and/or how, when and why the gene is expressed.

KEY CONCEPTS

- A test cross is used to determine whether an individual is homozygous dominant or heterozygous for a particular trait.
- A pedigree chart is used to trace the inheritance of a particular trait through several generations of a family.
- There are accepted symbols for constructing a pedigree chart.

CHECK YOUR UNDERSTANDING 5.2b

1 It is not known whether a Labrador dog is homozygous dominant for black coat or heterozygous.

- **a** Describe a cross that would enable you to find out.
- **b** What is this type of cross called?

2 A cross between peas homozygous for green pods and peas homozygous for yellow pods results in offspring that all have green pods.

- **a** Identify which gene is dominant and explain why.
- **b** If two of the resulting offspring with green pods are crossed, calculate the expected genotypic and phenotypic ratios. Show your working using either a Punnett square or a branching diagram.

3 Jordan has blue eyes. Both his parents are brown-eyed. He marries a brown-eyed woman, Claire, whose father has brown eyes and mother has blue eyes. Jordan and Claire have a child, Chloe, who has blue eyes.

- **a** State the genotypes of Jordan, Claire and Chloe.
- **b** Using a Punnett square, work out the chance of them having another blue-eyed child.
- **c** Draw a pedigree chart for the family.

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4 Use the information in Table 5.2 and Worked example 5.2 to help you to analyse the four pedigrees in Figure 5.16 and to solve the problems that follow.

- **a** State whether the trait being investigated in each pedigree (i–iv) is dominant or recessive, or whether the information given is insufficient to allow a valid deduction. Explain how you arrived at your conclusion for each.
- **b** Based on pedigree (ii), describe how individual II-2 is related to:
  - **a** individual II-1
  - **b** individual I-2.
- **c** Based on information given in pedigree (iii), use a Punnett square to determine the probability that the next child born would be homozygous recessive.
- **d** Based on pedigree (iv):
  - **a** Assign genotypes to each individual in the pedigree.
  - **b** Determine the probability that individual III-1 will be affected.
  - **c** Explain how individuals II-2 and II-3 are related.

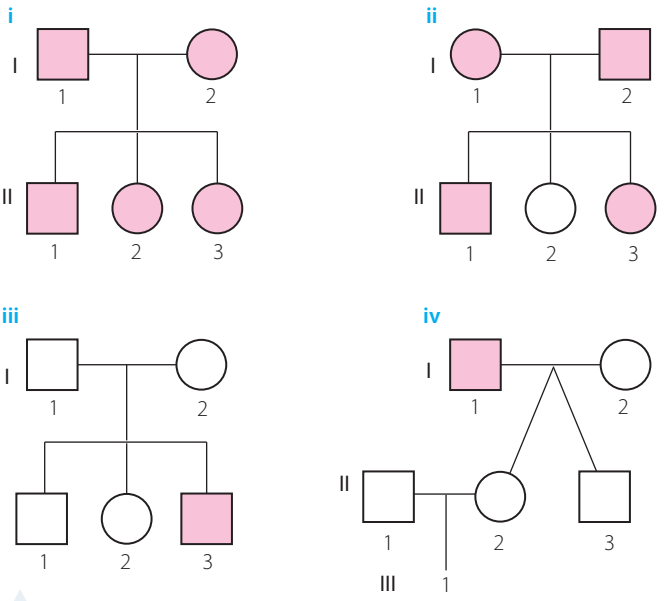


FIGURE 5.16 Pedigrees i – iv

5.3 Deviations from Mendel's ratios

Although Mendelian inheritance provides a solid model for inheritance patterns, later studies showed that at times there are deviations from Mendel's ratios under certain conditions. It is not known whether Mendel was aware that his ratios did not always hold true, or whether he chose to ignore results that did not fit in with his ratios.

Deviations from Mendelian ratios that have been observed over the years can be attributed to the following changes in typical Mendelian conditions:

- Some genes are not dominant or recessive; they may both be expressed (**codominance**) or a blending of their characteristics may be expressed (**incomplete dominance**).
- Some genes do not assort independently; they are linked. For example, genes on sex chromosomes show **sex-linked inheritance**, where ratios differ between male and female individuals.

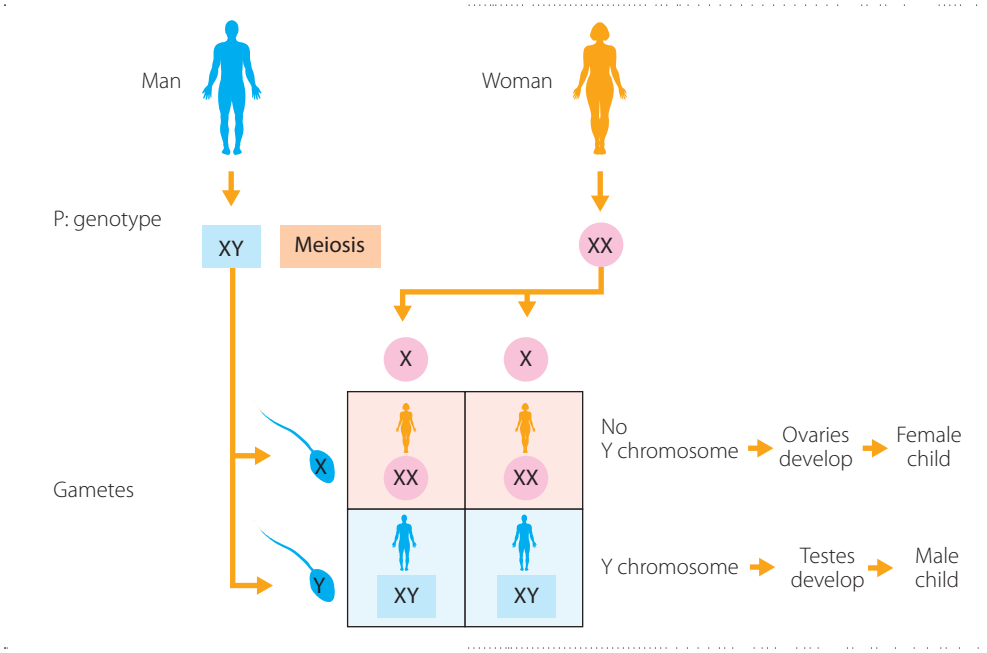
Sex determination

Sex determination is the way in which sex chromosomes separate during meiosis and then recombine during fertilisation to determine whether the offspring will be male or female.

During meiosis, the sex chromosomes segregate, just like any other homologous pair of chromosomes, where only one of each chromosome pair passes into a gamete. Because a female has 44 autosomes + XX, when the chromosome number is halved, each female gamete (egg cell) receives 22 autosomes + X. In males, half the gametes (sperm cells) receive 22 autosomes + X and the other half receive 22 autosomes + Y. Fertilisation follows and in the recombination of sex chromosomes in the zygote, the sex chromosome donated by the male gamete dictates the sex of the child (Fig. 5.17):

- A zygote that inherits an X chromosome from both the mother and the father will be female (XX).
- A zygote that receives an X chromosome from the mother and a Y chromosome from the father will be male (XY).

FIGURE 5.17 Sex determination in humans



In humans, genes on the X and Y chromosome code for the production of sexual reproductive organs and the development of secondary sexual characteristics that define whether an individual is phenotypically male or female. The Y chromosome carries the testis-determining gene and therefore, if present, the Y ensures that the child will be male. This does not apply to all animals.

Occasionally, **non-disjunction** of chromosomes occurs during meiosis, whereby the chromatids do not separate from each other. This results in an incorrect number of chromosomes in the offspring (for example, 45 or 47 instead of 46). (See Chapter 7 – Chromosomal mutations.) If this occurs in the sex chromosomes and only one X chromosome is present, the child will be female. An individual with XXY will be male (Chapter 15).

Weblink
Sex determination

Worksheet
Additional information on sex determination

Worksheet
Sex linkage diagrams and additional information on colour blindness

Worksheet
Summary of sex linkage in humans

Sex linkage

Sex linkage occurs when some genes carried on the X and Y chromosomes code for characteristics other than the gender of the individual. If a gene occurs on the X chromosome, females will have two alleles for that gene whereas a male will only have one, because he has only one X chromosome. Therefore recessive disorders appear more frequently in males.

For example, in humans, genes for red–green colour vision are carried on the X chromosome. The mutant form may result in the person being red–green colour blind (unable to distinguish between red and green). Haemophilia (a bleeding disorder) is an X-linked disorder. Alleles for this gene occur on the X chromosome. A male who inherits one copy of the mutant allele (on the X chromosome from his mother) will suffer from the condition. Because the male has no equivalent allele on the Y chromosome to mask this defective allele, a single copy of this recessive gene results in the male being affected by the recessive gene.

Females have two X chromosomes, one from each parent. Therefore if a female inherits a mutant allele for haemophilia on one X chromosome, she will not suffer from the disorder if her other allele is dominant. Such a female is termed a **carrier** – the defective allele does not affect her, but may be passed to her sons (who would be affected) or to her daughters (who may be carriers or affected, depending on the allele they inherit from their father). If a daughter inherits a pair of defective alleles for haemophilia (one from each parent, on each X chromosome), the condition is lethal. (Fig. 5.18 shows inheritance of haemophilia in the royal families of Europe.)

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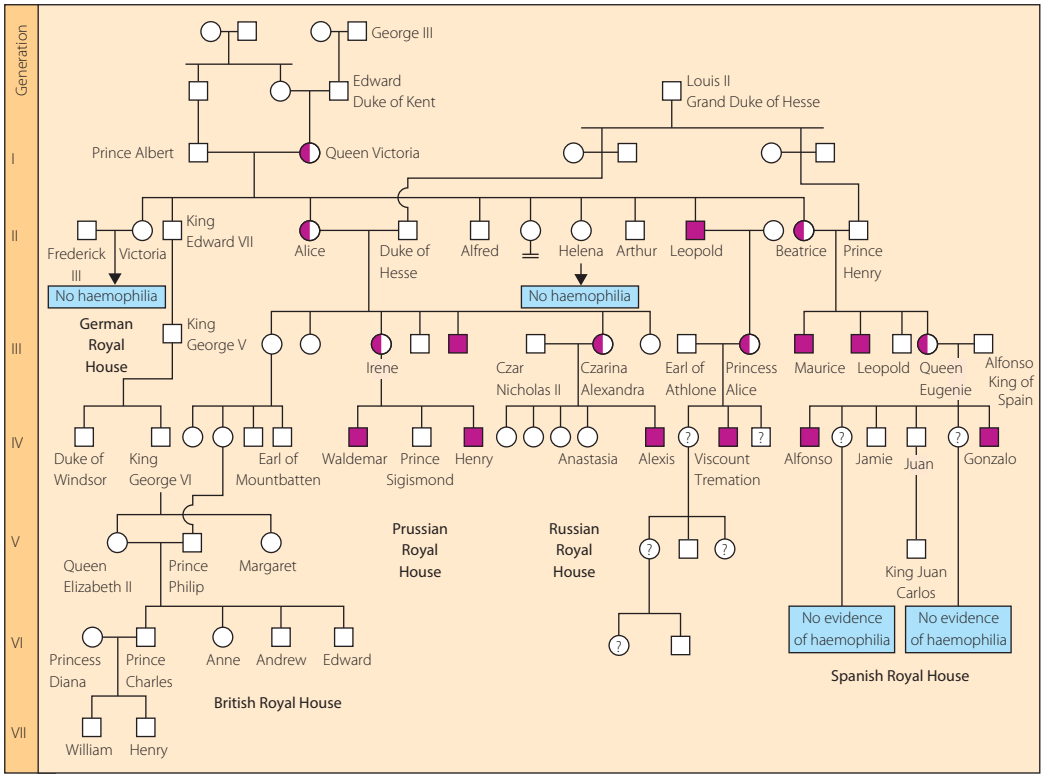
Some sex-linked genes are found on the Y chromosome (termed 'Y-linked') and appear in males only, but these tend to be less common. An example of a Y-linked gene is Y chromosome infertility.

a



b

FIGURE 5.18 **a** Pedigree showing sex-linked haemophilia in the British Royal family; **b** Queen Victoria and her family, who were affected by this disease

Symbols used to represent alleles in sex-linked crosses

When the inheritance of a *sex-linked* gene is being represented, the *alleles* of that gene (for example, H = normal blood clotting, h = haemophilia) as well as the *type of chromosome* on which it is carried (X or Y) must be shown in the genetic cross. The letters H and h are assigned in keeping with the symbols used to show dominant and recessive alleles of a gene. These are then written as a superscript on the chromosome on which they are attached.

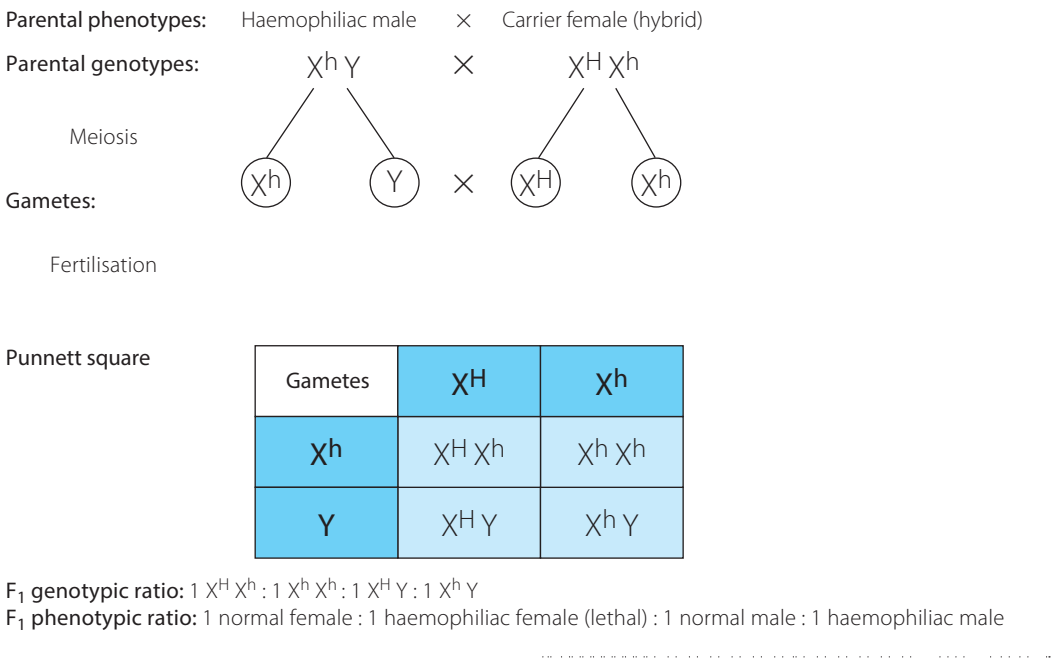
For example:

$X^H X^H$ = normal female
$X^H X^h$ = carrier female (heterozygous)
$X^h X^h$ = haemophiliac female (homozygous = lethal)

$X^H Y$ = normal male
$X^h Y$ = haemophiliac male

Segregation of both the chromosome and the attached alleles can therefore be represented (Fig. 5.19).

FIGURE 5.19
Genetic cross of two hybrid parents for the sex-linked trait haemophilia



Punnett square

Gametes	X^H	X^h
X^h	$X^H X^h$	$X^h X^h$
Y	$X^H Y$	$X^h Y$

F_1 genotypic ratio: 1 $X^H X^h$: 1 $X^h X^h$: 1 $X^H Y$: 1 $X^h Y$
F_1 phenotypic ratio: 1 normal female : 1 haemophiliac female (lethal) : 1 normal male : 1 haemophiliac male

Weblink
Colour vision
Read the information on the types of colour blindness, and explore pedigrees of inheritance.

The results of this sex-linked genetic cross can be analysed as follows:

- The ratio of males to females = 1 : 1
- Looking at the males only, there is a phenotypic ratio of *1 normal male : 1 affected male*.
- There is a 25% probability that any offspring will be affected by the disease. There is a 50% probability that a *boy* is affected.

Not all sex-linked disorders are lethal in homozygous recessive females. Examples where the homozygous recessive is not lethal is red–green colour blindness in humans and white eye colour in fruit flies.

Weblink
Colour vision tests and charts

Incomplete dominance and codominance

Weblink
Sex-linked inheritance online problems

Incomplete dominance and codominance are two more examples of inheritance that does not show a Mendelian pattern. This is because in the genes of some organisms, pairs of alleles do not show dominance of one allele over the other.

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Incomplete dominance

In incomplete dominance there is a *blending* of the features of the two alleles expressed, giving a hybrid that is intermediate; for example, red snap dragon flowers crossed with white snapdragon flowers give pink flowers. Incomplete dominant pink hybrids are also seen in tulips, carnations and roses.

Special notation is used to represent alleles that do not show complete dominance. A letter is chosen to represent the gene – for example, C for colour. The alleles are written as superscripts next to the gene, so the allele for red would be C^R and the allele for white would be C^r (Fig. 5.20).

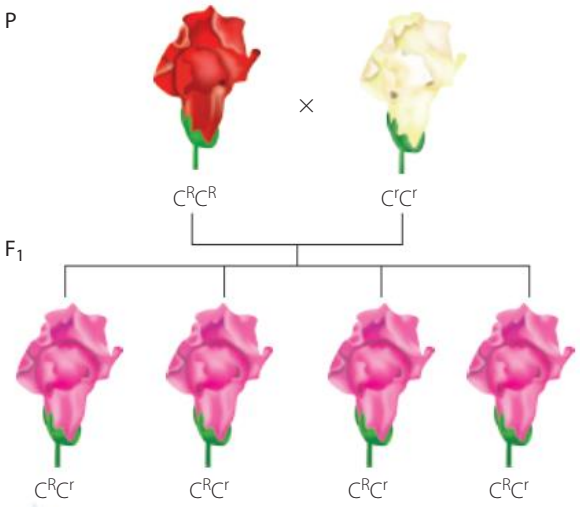

FIGURE 5.20 Incomplete dominance in snapdragon flowers

Codominance in animals

In codominance *both alleles* are expressed, creating a new phenotype. The term 'codominant' describes this (*co* = together; *both* alleles behave as *dominant* alleles because they are both expressed).

Pure-breeding (homozygous) cattle may have a red or white coat colour. Hybrid individuals (heterozygotes), which have one allele for red and one for white coat colour, have a roan appearance – both red and white hairs are present, not in patches but interspersed. That is, both colours of hair are present, indicating that both alleles are expressed, a typical example of codominance (Fig. 5.21).

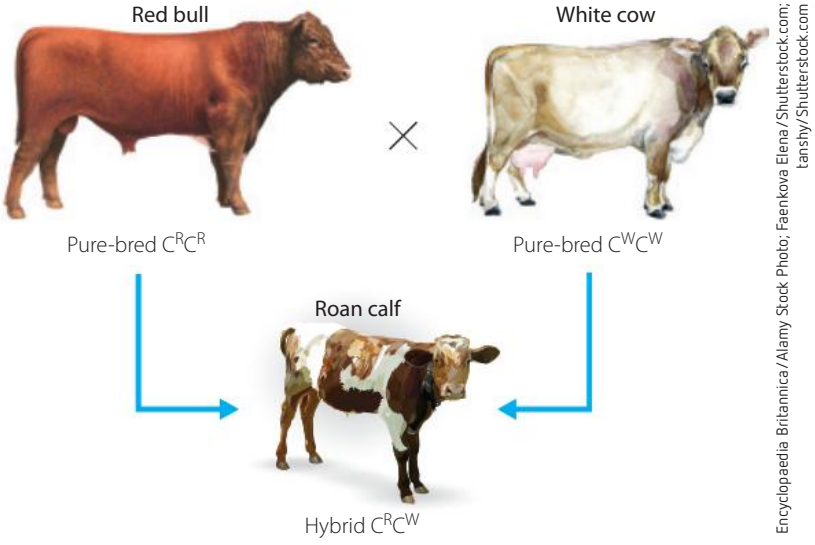

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FIGURE 5.21 Codominance in cattle: a red bull and a white cow produce a roan calf. Close inspection of the roan calf reveals a mottled appearance of red and white interspersed hairs.

Weblink
Incomplete dominance and codominance

Another example of codominance occurs in Andalusian chickens. If a homozygous black fowl is crossed with a homozygous white fowl, the heterozygous offspring in the F_1 generation appear 'blue'. At first this was thought to be a 'blending' of characteristics (incomplete dominance), but closer examination revealed that the blue Andalusian fowls have both black and white feathers present – a typical example of codominance.

Special genetic notation is used to represent codominance. A letter is chosen to represent the gene – for example, C for colour. The alleles are written as superscripts next to the gene. Because both alleles are expressed, each allele is given a capital letter. Therefore red would be C^R and white would be C^W. The roan cow would be represented as C^RC^W.

Worksheet
Deviations from simple Mendelian ratios

KEY CONCEPTS

TABLE 5.3 Comparison between autosomal recessive inheritance and other inheritance patterns

CONDITIONS FOR AUTOSOMAL RECESSIVE INHERITANCE (MENDELIAN RATIOS)	TYPE(S) OF INHERITANCE AND CONDITIONS THAT CAUSE VARIATION FROM THE MENDELIAN INHERITANCE
Individuals have two alleles for each characteristic; alleles may be the same (such as in pure-breeding individuals) or they may differ (as in hybrid individuals).	*Sex-linkage:* the heterogametic sex may have only one factor. For example, in humans, males have one X and one Y chromosome (and the Y is much smaller than the X because it lacks some of the genes present on the X). The male genotype is XY and so X-linked genes are absent from the Y chromosome, and therefore only one copy is present.
Genes are inherited as 'discrete units' and are not dependent on whether they come from the male or female parent.	*Sex-linkage:* some genes coding for non-sexual characteristics are located on the sex chromosomes. These show inheritance patterns similar to the sex chromosomes on which they occur. For example, the sex-linked recessive trait for colour-blindness, present on the X chromosome and not on the Y, appears more frequently in the phenotype of males because there is no paired allele (no second X chromosome) to mask its effect.
The trait that is expressed in hybrid individuals is *dominant*, and the trait that is hidden or masked is the recessive trait (Mendel's first law – dominance).	*Codominance:* both alleles in the hybrid are expressed, and so one allele is not dominant to another (e.g. roan cattle, which are hybrids resulting from cross-breeding a red parent with a white, have both red and white hairs). *Incomplete dominance:* both alleles are expressed as a blending of the two characteristics (e.g. red flowers crossed with white flowers give offspring with pink flowers).
When two hybrids breed, statistically they will produce a ratio of 3 : 1 offspring – *three* offspring showing the same trait as the parents (termed the *dominant* trait) to *one* offspring showing the contrasting *recessive* trait.	Ratios change for both sex-linked and codominant genes: • *Sex-linkage:* any recessive genes on the X chromosome in males will be expressed in the male phenotype of offspring, because they are unpaired (there is no equivalent dominant gene present). • *Codominance* and *incomplete dominance:* an offspring that is hybrid does not resemble either pure-breeding parent. It has a different phenotype of its own, as the genes of *both* parents are expressed in the individual (codominance) or a blending of the parental genes is expressed (incomplete dominance). Therefore the ratio of a monohybrid cross will be 1 : 2 : 1.

CHECK YOUR UNDERSTANDING 5.3a

1. For each of the following figures, draw a Punnett square to represent the cross. Give the expected genotypic and phenotypic ratios, and explain the type of inheritance pattern shown.
   - **a** Figure 5.9 (one Punnett square for each cross)
   - **b** Figure 5.12a × Figure 5.12b
   - **c** Figure 5.14
   - **d** Figure 5.20
   - **e** F1 cross in Figure 5.21
2. Draw a pedigree chart to show the trait *roan colour* in cattle, using Figure 5.21. Assume that the roan calf in the figure is male. Add to the pedigree, showing the next generation where the roan calf, once mature, mates with a white cow, giving rise to twin male calves (one roan and one red) and to a white female calf.
3. Distinguish between 'codominance' and 'incomplete dominance'.
4. If a white chicken is mated with a brown chicken, the offspring have both brown and white feathers.
   - **a** Identify the type of inheritance.
   - **b** Use a Punnett square to predict the expected genotypes and phenotypes of the offspring if two of the hybrid offspring are hatched.
5. Explain why genes that are sex-linked do not give Mendel's predicted ratios when individuals are crossed.

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Multiple alleles

Individuals usually have only two alleles for each gene (or one, in the case of sex-linkage). Within a population, however, there may be three or more alleles for a single gene trait. Such a trait is termed **multi-allelic**. For example, in humans, the gene for human blood type has three alleles in the population: A, B and O.

Blood groups are one of the most commonly studied genetic variations in human populations. This is mainly because transfusions of the wrong type of blood can lead to death. Blood cells have molecular markers on their surfaces and these play an important role in allowing a person's own body cells to be recognised by the immune system as 'self' (that is, cells belonging to that individual).

To represent multi-allelic blood groups using correct genetic notation, the gene is denoted as I and the three alleles are represented by superscripts: I^A, I^B and I^i. Alleles A and B are codominant, as they each produce a molecular marker on red blood cells. If both alleles are present, the blood cells have both markers. The i allele produces no molecular marker on the red blood cells and is recessive to both A and B. As a result, there are four possible phenotypes for the ABO blood system: a person may have blood group A, B, AB or O. There are, however, six possible genotypes (Fig. 5.22, Table 5.4).

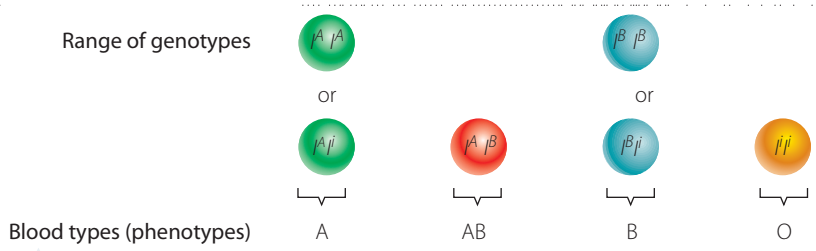

FIGURE 5.22 ABO blood types in humans – a range of genotypes showing multi-allelic inheritance

TABLE 5.4 Human blood groups: genotypes and phenotypes

ALLELES	GENOTYPE	MOLECULAR MARKERS PRESENT	PHENOTYPE (BLOOD GROUP)	DOMINANCE OR CODOMINANCE
AA	Homozygous	A	A	—
BB	Homozygous	B	B	—
AB	Heterozygous	AB	AB	Codominance
AO	Heterozygous	A	A	Dominance
BO	Heterozygous	B	B	Dominance
OO	Homozygous	O	O	—

Another example of a gene that has multiple alleles is the gene for coat colour in rabbits. There are four alleles, called *normal*, *chinchilla*, *himalayan* and *albino*.

It is important not to confuse a phenotypic trait that has *multiple alleles* with phenotypic traits that are coded for by *multiple genes*. For example, height and eye colour in humans are termed **polygenic** traits as they have two or more genes coding for them and each gene has its own set of alleles. As an example, height in humans shows **continuous variation**, with a smoothly graded range of heights from short to tall, the result of many genes coding for the single trait (human height). This is in contrast to the single-gene inheritance of Mendel's garden peas, where there are only two alleles for height – tall or short (**discontinuous variation**).

INVESTIGATION 5.3

Modelling inheritance patterns

Weblink
Video: Genetics in everyday life.
Make notes to clarify your understanding of inheritance patterns.

Weblink
Making Reebops
Review the model on the website as a starting point for designing your own model to explain the different types of inheritance patterns.

Weblink
Conduct your own virtual experiments with fruit flies

Weblink
Explore dihybrid crosses

You are required to model inheritance patterns, showing the formation of new combinations of genotypes produced during meiosis. The inheritance patterns you are required to model include autosomal inheritance, sex-linkage, codominance, incomplete dominance and inheritance of multiple alleles.

Use pairs of homologous chromosomes on which you mark the different types of alleles (Fig. 5.23).

In your model, include the process of fertilisation to predict how variation arises in genotypes of offspring.

RESOURCES

Use the weblinks to design your model. You may wish to create your model using your own version of 'Reebop' genes – see weblink – or genes in a model of your own design, to explain each of the inheritance patterns listed below.

PART A: INHERITANCE PATTERNS

Model the following inheritance patterns passed from parents to the F_1 generation:

A: autosomal inheritance

B: sex-linkage

C: codominance

D: incomplete dominance

E: multiple alleles.

1 Take photographs of the phenotypes of the parents you use and of the offspring that you create. Record the genotypes and phenotypes in a table.

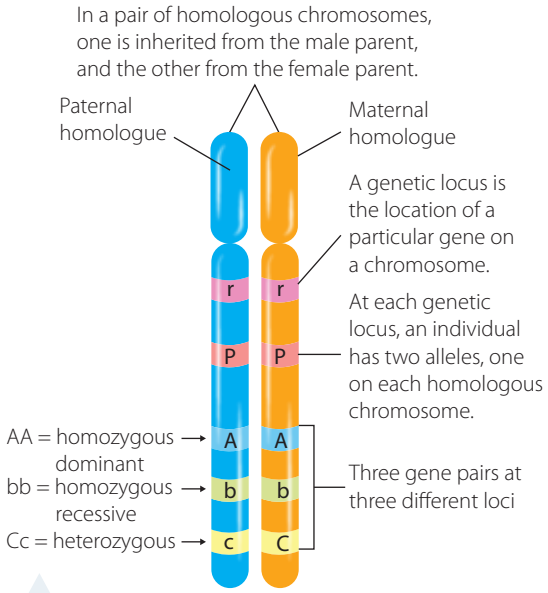

FIGURE 5.23 Homologous chromosomes with pairs of alleles showing possible combinations

PART B: PUNNETT SQUARES AND PEDIGREES

2 Using your model, create Punnett squares to explain each inheritance pattern (A to E) that you have modelled. (You may wish to divide this up among the members of the group so that each member creates one or two Punnett squares.)

3 Create a pedigree chart for each inheritance pattern A to E, to show inheritance of genes from parents to F_1 as modelled. Then continue the pedigree chart to predict the possible types of offspring if F_1 are crossed to produce the F_2 generation.

4 Work collaboratively as a group to present your results in a table.

EXTENSION

Use the weblinks to conduct virtual experiments online using fruit flies, or explore inheritance of two or more genes (dihybrid crosses).

KEY CONCEPTS

- In a multiple allele system, one gene has three or more alleles present in the population – an example is the ABO blood group system.
- A and B alleles are codominant in the heterozygous form, as they are both expressed.
- The allele for O blood type is recessive to both alleles A and B.
- Multiple alleles are many different versions of the same gene, whereas polygenic traits have many genes that determine one trait.

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CHECK YOUR UNDERSTANDING 5.3b

1 Define 'sex-linked trait'.
2 Write the correct genetic notation to show ABO blood groups.
3 Using the ABO blood system as an example, draw a Punnett square to show a genetic cross that can produce all four possible phenotypes in this inheritance. Use examples from this to explain the difference between dominant and codominant alleles.
4 Distinguish between multiple alleles and polygenic traits.
5 Write three genetic problems for a classmate to answer. The questions should target three different inheritance patterns.

5.4 Population genetics

The genetic similarities and differences within and between species may be determined at the phenotype, genotype, allele or molecular level. To do this, scientists conduct frequency studies. They gather quantitative data that can be analysed and applied to understand variation in individuals, populations and whole species. Scientists use this data to predict the potential of populations to adapt, as well as the future resilience and survival of the species.

Population genetics is the study of how the gene pool of a population changes over time, leading to a species evolving. It relates directly to the inquiry question at the start of this chapter, which asks how the genetic similarities and differences within and between species can be compared.

The **gene pool** is the sum total of all the genes and their alleles within a population. **Genetic diversity** is the total of all the genetic characteristics in the genetic makeup of a species. It is dependent on **genetic variability**, the tendency of individual genetic traits in a population to vary. Species that have a greater degree of genetic diversity have a greater potential to adapt and survive.

Review *Biology in Focus Year 11*, Chapter 9, on macroevolution and microevolution.

Population genetics combines the concepts of Mendelian genetics and Darwinian evolution to explain how changes in allele frequencies arise in populations and how these changes can lead to microevolution (over a relatively short period of time) and macroevolution (over a long period of time).

Populations differ in the extent of their genetic variation for particular genes (Fig. 5.24). By measuring the degree of genetic variation within a population over time, scientists can make predictions about how populations adapt to their environments and which populations are more likely to flourish, evolve into new species or die out.

Population geneticists study factors that cause changes in allele frequency within a population. For example, they may investigate how a temperature change (selective pressure) affects the allele frequency in a particular population. They use a model based on allele frequencies typical of a stable population with Mendelian inheritance (a population in equilibrium) and then compare this with allele frequencies in a real population exposed to selective pressures. External factors other than natural selection that cause changes to allele frequency are studied, such as *gene flow* and *genetic drift*, which you will learn about in Chapters 6 and 7.

To conduct a scientifically valid study, population geneticists gather quantitative data – they measure gene and allele frequencies within real populations – and then apply an abstract mathematical model to predict how external factors will influence these frequencies. They test their conclusions against empirical data (data obtained by observation and experimentation) so that they can draw valid conclusions about patterns of genetic variation within populations over time.

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FIGURE 5.24 The frequencies of characteristics in populations change over time.

Genetic variation and frequencies of characteristics

As you know, genetic variability within a population is essential for evolution by natural selection. Microevolution can be studied by examining a change in the frequency of alleles in a population over several generations. For example, skin colour in the red-eyed tree frog *Agalychnis callidryas* is determined by a gene that has two alleles: A (normal colour) and a (albino) (Fig. 5.25).

Genetic variability in a population can be determined by analysing the relative proportion (ratio or percentage) of a given phenotype, genotype or allele within that population. The discussion of genetic crosses using Punnett squares, earlier in this chapter, looked at the different phenotypic and genotypic ratios in offspring. Another important factor is allele frequency in a population.

Allele frequency is a measure of how common an allele is within a population. Many genes are **bi-allelic** – that is, they have two variants or two possible alleles within a population (for example, T and t for height of pea plants). Allele frequency can be calculated by counting the number of copies of an allele in a population and then dividing by the total number of copies of all alleles of the gene:

$$\text{Frequency of allele G} = \frac{\text{Number of copies of } \textit{allele} \text{ (G) in the population}}{\text{Total number of copies of the } \textit{gene} \text{ (G + g) in the population}}$$

Some genes may be multi-allelic (more than two allele variants per gene) – an example is the gene for blood group. In this case, to calculate the total number of copies of the gene, we would need to add together all the different alleles (three for ABO blood groups) within the population.

worldswildlifewonders/Shutterstock.com

MARK SMITH/Science Photo Library (SPL)

FIGURE 5.25 Skin colour in the red-eyed tree frog. Normal colour (A) is dominant over albino (a).

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WORKED EXAMPLE 5.3

If you conduct a study of a small population of tree frogs over time, you can measure the change in allele frequency. Tables 5.5a and 5.5b represent a hypothetical frog population, showing phenotypes present in the first generation (Table 5.5a) and seven generations later (Table 5.5b).

1 For each generation of the frog population, calculate the:
   - **a** phenotypic frequencies
   - **b** genotypic frequencies
   - **c** allele frequencies.

2 Propose possible reasons for the change in allele frequencies.

3 Propose a research question or hypothesis that you as a population geneticist may decide to test, to find out whether a particular factor is the basis for the change in allele frequency.

TABLE 5.5a Frog population in first generation (generation 1)

 AA	 Aa	 AA	 aa
 AA	 AA	 AA	 aa
 AA	 Aa	 AA	 Aa

TABLE 5.5b Frog population in generation 7

 aa	 Aa	 Aa	 aa
 AA	 AA	 Aa	 aa
 AA	 Aa	 aa	 Aa

ANSWER			LOGIC
Generation:	First (1)	Later (7)	
(i) *Phenotypic frequencies:*			▪ Count the number of normal and albino frogs in each generation. ▪ Calculate the phenotypic frequencies by dividing the number of each type of frog by the total number of frogs. Round to two decimal places.
Normal frogs	$\frac{10}{12} = 0.83$	$\frac{8}{12} = 0.67$	
Albino frogs	$\frac{2}{12} = 0.17$	$\frac{4}{12} = 0.33$	
(ii) *Genotypic frequencies:*			▪ Count the number of each type of genotype (AA, Aa and aa) in each generation. ▪ Calculate the genotypic frequencies by dividing the number of each type of genotype by the total number of frogs. Round to two decimal places.
AA	$\frac{7}{12} = 0.58$	$\frac{3}{12} = 0.25$	
Aa	$\frac{3}{12} = 0.25$	$\frac{5}{12} = 0.41$	
aa	$\frac{2}{12} = 0.17$	$\frac{4}{12} = 0.33$	
(iii) *Allele frequencies:*			▪ Count the number of each type of allele (A and a) in each generation. ▪ Calculate the allele frequencies by dividing the number of each type of allele by the total number of alleles. Round to two decimal places.
A	$\frac{17}{24} = 0.71$	$\frac{11}{24} = 0.46$	
a	$\frac{7}{24} = 0.29$	$\frac{13}{24} = 0.54$	
Possible reasons for change	Yellow colour has a selective advantage. More yellow frogs moved into the population.		▪ Think of as many reasons as possible with a genetic and/or evolutionary basis.
Thinking	I wonder if the colour affects which frogs are captured by predators.		▪ State what you are thinking/wondering.
Research question	What effect does the albino colour of frogs have on capture by predators?		▪ Narrow this down to a more specific research question.
Hypothesis	If a frog is albino, then its rate of capture by predators will be less frequent.		▪ Turn the research question into a hypothesis.

TRY THESE YOURSELF

1 Calculate genotype, phenotype and allele frequencies for this population of peppered moths over three generations. Assume black moths are homozygous dominant. Give reasons for the possible change in colour and write a hypothesis that could be tested experimentally.

Flickr / Ben Sale; iStock.com / Henrik_L

FIGURE 5.26 Change in peppered moth colour over three generations

2 Calculate the phenotypic, genotypic and allele frequencies for the ABO blood groups in the family shown in the pedigree (Fig. 5.27). Their genotypes are as follows: father AO, mother BO, son AB, daughter AO, baby O.

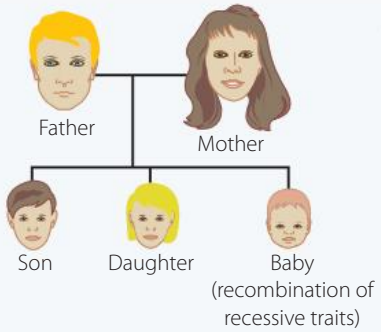

FIGURE 5.27

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INVESTIGATION 5.4

Examining frequency data in genetic studies

Numeracy

Critical and creative thinking

There are many ways to answer common questions on genetics, such as:

'What is the probability that an individual will inherit the genetic trait *x*?'

'Is it likely that population *y* will be able to adapt and survive as a result of a change in the environment?'

'Is gene *z* more common in people of particular ethnic groups, or those living in certain countries in the world?'

To make accurate predictions and provide answers to these and other questions relating to population genetics, quantitative frequency data is used.

PART A: INTERPRETING FREQUENCY DATA FROM GRAPHS

Examining the frequencies of blood groups of humans around the globe shows differences between people from different countries and ethnic origins.

Analyse the data in the graph in Figure 5.28 and identify any trends, patterns and relationships that are evident.

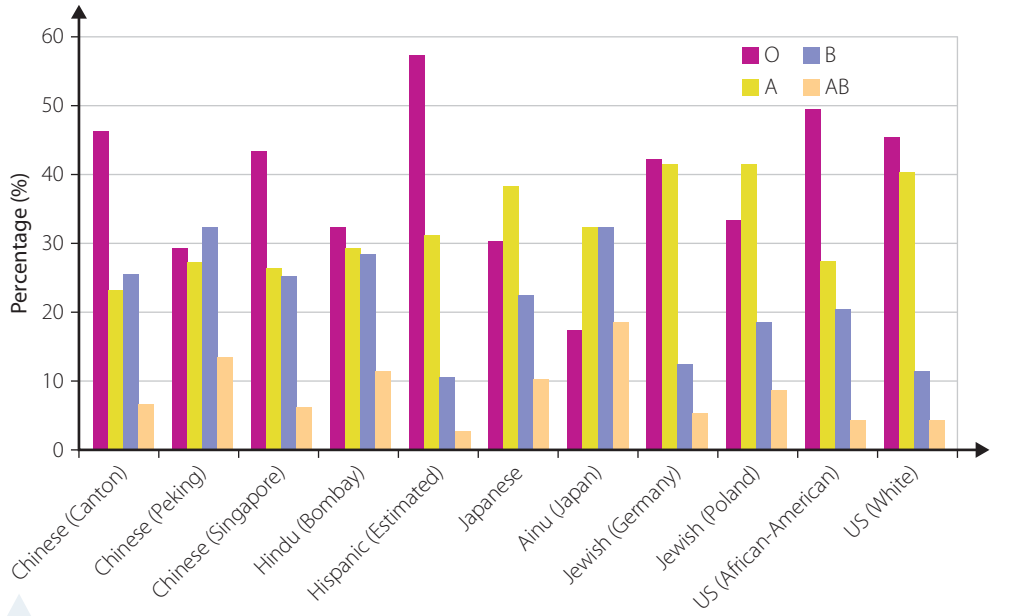

FIGURE 5.28 Frequencies of blood groups of humans around the world

1. Does the frequency data represent phenotype, genotype or allele frequencies? Justify your answer.
2. State which blood types are most and least common among most groups of people.
3. Which two groups differ most significantly in their blood group frequencies compared with groups from the rest of the world?
4. Calculate the average frequency of each blood group in all sectors of people represented in the graph.
5. Draw two pie graphs to represent the blood group distribution of Japanese and Ainu Japanese people. Construct a table to compare the distribution of blood groups in these two groups.
6. Formulate three questions of your own – two that can be answered from the graph and one that you find interesting and would like to research further.
7. Conduct a secondary-source investigation and write a paragraph to answer the research question that you formulated.

»

Weblink
Analysis of gene frequency of two alleles in a population of wild rabbits
Calculate the gene frequency of the alleles for each generation, and then graph the frequency of the two alleles over 10 generations.

8 Pair with another student and each answer the other's questions related to the graph. Share your findings on your researched questions.

9 Brainstorm with your partner as many reasons as you can to explain why the frequency data represented in Figure 5.28 would have relevance for the Red Cross. (The international Red Cross is a global organisation that helps with blood donations during conflict, in response to human-made or natural disasters or due to conditions of chronic poverty.)

PART B

1 Research why the frequency of the gene for sickle cell anaemia, a detrimental mutation that affects red blood cells, has a higher frequency in African people than in other populations. Find a graph to support this claim.

2 Extension: Model gene frequency change in a population affected by natural selection (evolution). Conduct an investigation using allele frequency, as outlined in the weblink.

KEY CONCEPTS

- Population genetics combines Mendelian genetics and Darwinian evolution to explain how changes in allele frequencies arise in populations and how these changes can lead to microevolution and macroevolution.
- Population geneticists study mathematical changes in gene and allele frequencies in populations to develop quantitative ways of exploring different evolutionary hypotheses.
- Allele frequency is a measure of how common an allele is in a population:

$$\text{Frequency of allele G} = \frac{\text{Number of copies of } \textit{allele} \text{ (G) in the population}}{\text{Total number of copies of the } \textit{gene} \text{ (G + g) in the population}}$$

CHECK YOUR UNDERSTANDING 5.4a

1 What is population genetics?

2 How do population geneticists gather, process and apply data to make predictions about populations?

3 Write out a mathematical formula for calculating:
- **a** phenotypic frequency
- **b** genotypic frequency
- **c** allele frequency.

4 Give one application for gathering each of the types of data listed in Question 3.

Mutations are dealt with in more detail in Chapter 7.

Single nucleotide polymorphisms

In genetics, the term **polymorphism** refers to individuals with different phenotypes. Literally translated, it means 'many forms' (*poly* = many, *morph* = form). Polymorphisms usually arise as a result of a mutation – an error in DNA replication.

A single nucleotide polymorphism or SNP is pronounced 'snip'.

A **single nucleotide polymorphism** (SNP) is like a typing error in DNA, where one nucleotide is replaced by another (Fig. 5.29). SNPs usually arise during DNA replication, where a single nucleotide is incorrectly inserted, creating an error in the DNA sequence at a particular location on a chromosome. To be termed a SNP (rather than simply a mutation), this altered DNA sequence must occur in at least one per cent of the population.

Why are SNPs useful in genetic studies?

Variations in organisms, including SNPs, may be associated with phenotypic change, such as a change in appearance, enzyme functioning, disease susceptibility or response to drugs. Most SNPs, however,

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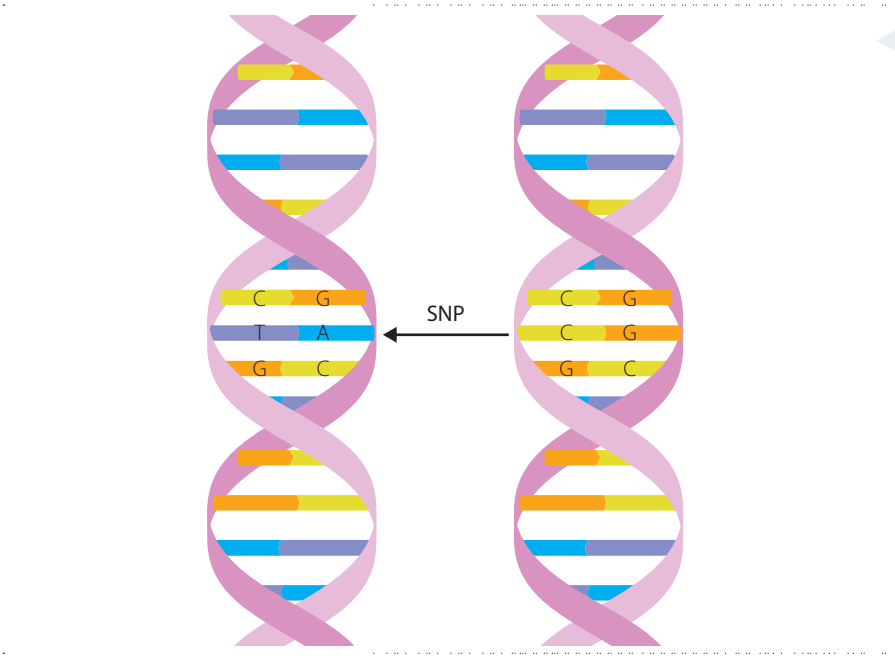

FIGURE 5.29 Single nucleotide polymorphism (SNP)

occur in non-coding regions of DNA and do not lead to observable differences. What, then, is the importance of studying these SNPs?

SNPs are important genetic markers that are currently used to distinguish individuals and to identify things such as disease susceptibility in individuals. A **genetic marker** can be defined as an identified sequence of DNA at a known site (locus) on a chromosome – for example, a SNP or a **STR (short tandem repeat)**.

Individuals within a population show great variation in the genetic markers they have on their DNA. This gives scientists a relatively easy way to tell individuals apart. For example, there are approximately 10 million known SNPs in the human genome.

Some genetic markers are associated with specific traits or disorders, but do not necessarily cause them. In studies of genetic markers, called **genome-wide association studies (GWAS)**, computer technology is used to rapidly scan genetic markers across the genomes (complete sets of DNA) of many people to find genetic variations associated with a particular disease. A data bank of SNP genetic markers is currently being built to record associations between the presence of specific markers and particular diseases or disorders.

Progress in DNA manipulation techniques and advances in bioinformatics technology (computer analysis of biological data) has allowed very large numbers of SNP markers to be identified in particular regions of chromosomes. Genome-wide association studies are based on the presence of a *group* of SNP markers (called a **haplotype**) associated with a trait, rather than trying to link an *individual* SNP with a trait. Some applications of identifying haplotypes are:

- as indicators of disease
- to establish family lineage and determine the genetic relatedness of individuals
- to study evolutionary relatedness (Chapter 6).

Frequency of SNPs and genome-wide association studies

On average, the frequency of SNPs is approximately one in every 300 nucleotides in the human genome, giving a total of approximately 10 million SNPs, most of which occur in the non-coding regions (introns) of the DNA.

Weblink
What are SNPs?

You will learn more about STRs in Chapter 6.

You will learn about the application of haplotype studies in Chapter 6.

Weblink
Making SNPs make sense
SNP data can be applied, for example, to choosing prescription drugs.

In genome-wide association studies (GWAS), scientists look for SNPs that occur in higher frequencies in people with a particular disease, compared with people who do not have the disease. The SNPs that occur in a higher frequency are said to be *associated* with that disease. Advances in technology allow hundreds or thousands of SNPs to be analysed at the same time, and this is much faster and cheaper than sequencing whole genomes. By studying SNPs in groups of 25–50 people, scientists are able to detect polymorphisms occurring in 1 to 3 per cent of the population with approximately 95 per cent confidence. (See Chapter 1, Accuracy, precision and errors in measurement.)

Data from studies such as these are entered into data banks that other scientists and researchers can access. Haplotype analysis to date has shown an association between particular SNPs and human diseases, such as osteoporosis, asthma, diabetes and Alzheimer's, to mention only a few. The HapMap project (short for 'haplotype mapping') is an international project that involves scientists collectively cataloguing markers inherited together on chromosomes, including markers such as SNPs) and short tandem repeats (STRs), to create databases that can be searched to examine these common genetic variants in detail. Over the past decade, data such as this has been gathered on an enormous scale.

Limitations of SNP data

Scientists have found that many biological questions can be answered using smaller regions of the genome that show polymorphisms. This data is reliable as long as the regions selected are fairly evenly distributed throughout the genome. Selection of markers is also important – genetic markers that are closer together give more accurate data. This is because, for a haplotype study, one must look at SNPs that have been inherited from one parent. If there is crossing over during meiosis, the SNPs on a chromosome might not all be inherited together. Keeping these and other more technical limitations in mind, scientists are using SNP data more frequently.

Why use SNPs rather than whole-genome DNA sequencing?

The terms *genotyping* and *sequencing* have slightly different meanings. **Genotyping** involves identifying genetic variations in individuals, whereas **sequencing** involves finding the exact nucleotide sequence (in terms of A,T, G and C) of a certain length of DNA (Fig. 5.30). A whole genome, parts of the genome or a short piece of DNA can be sequenced.

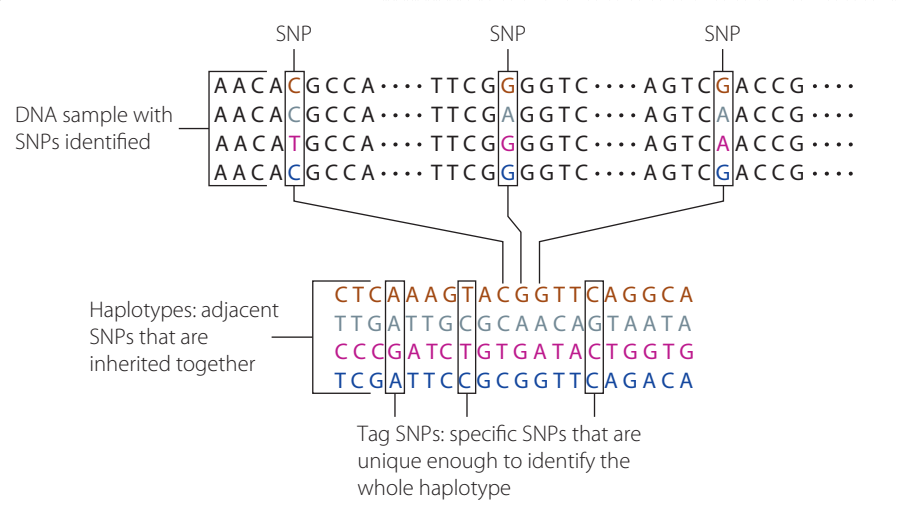

FIGURE 5.30 DNA sequencing information showing SNP changes in DNA

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INVESTIGATION 5.5

A secondary source investigation of how genetic similarities and differences can be compared

PART A: ANSWERING THE INQUIRY QUESTION AT THE START OF THIS CHAPTER

Answer the inquiry question at the start of this chapter, recording your information in the form of a concept map.

How can genetic similarities and differences within and between species be compared?
Put this question in the centre of your concept map.

Review the variety of methods studied in this chapter, from investigations using genetic crosses, through studies of allele frequency data, to using SNPs, to explain how genetic similarities and differences within and between populations can be compared.

As a starting point, consider which methods are useful for finding genetic similarities and differences:

- between individuals
- within populations
- between species.

Leave space on your concept map, as you might want to add details after you have completed chapters 8 and 9.

PART B: EXTENSION – INVESTIGATING SNPS AND THEIR APPLICATIONS

1. See the worksheet 'Extension: Understanding SNP data'.
2. Investigate what a SNP is and what SNP data is. Find out how SNP data differs from DNA sequencing data, what the applications (uses) of SNP data are, and any limitations of this type of data.
3. Look up PheGenI (Phenotype-Genotype Integrator) or other software programs and find a YouTube clip less than four minutes long that explains and demonstrates how SNP data software works.

Worksheet
Extension: Understanding SNP data

KEY CONCEPTS

- SNPs are loci on chromosomes where alleles differ at a single base. The rarer allele must have a frequency of at least 1% in a random set of individuals in a population to be termed a SNP.
- SNPS are valuable as genetic markers for identifying individuals and can be associated with certain disorders and traits.
- A haplotype is a cluster of marker alleles on the same chromosome and can be used for association studies in disease and to track the inheritance of different regions of the genome.

CHECK YOUR UNDERSTANDING 5.4b

1. Explain what a SNP is.
2. Give two advantages of using SNP techniques rather than whole-genome sequencing.
3. Describe three applications of SNP data.
4. How can the genetic similarities and differences within and between species be compared?
5. Go back to the concept map you created in Investigation 5.6 and fill in what you have learned.

5 CHAPTER SUMMARY

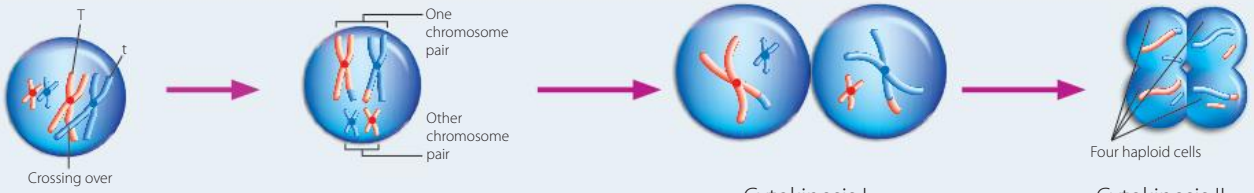
How can the genetic similarities and differences within and between species be compared?

HOW GENETIC VARIATION IS INTRODUCED

Meiosis

Crossing over | **Independent assortment** | **Halving of chromosome number** | **Four gametes, all genetically different**

Half of the gametes will contain T and half will contain t.

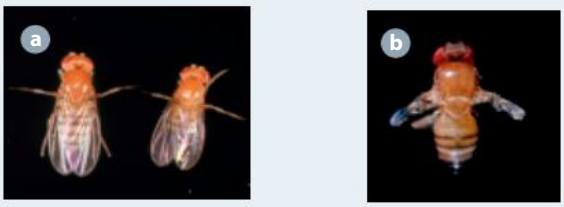


Early prophase I | Metaphase I Pairs of | Cytokinesis I | Cytokinesis II

Chromosomes align independently – various *combinations of paternal and maternal genes* are possible.

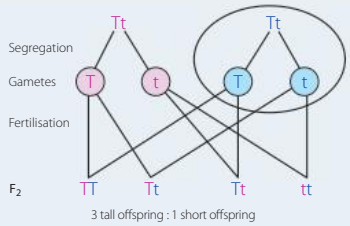
The genes in each haploid cell are a *combination of the parental genes* = variation is introduced.

Mutation



A mutation is a change in the DNA of a cell and may *give rise to new alleles* (different forms of the same gene) that produce different phenotypes (variation).

Fertilisation

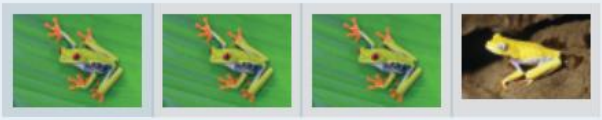


Two haploid gametes fuse. Chromosomes in the zygote have a *combination of the genes* contributed by the parents.

POPULATION GENETICS

Allele frequency

Population genetics studies how the ratios of alleles in the gene pool of a population change over time. It is a way *of comparing genetic similarities and differences* within and between species.

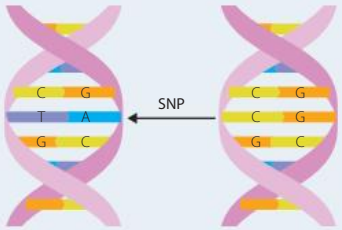


Using the formula one can *calculate allele frequency*:

$$\frac{\text{Number of copies of } \textit{allele} \text{ (G) in the population}}{\text{Total number of copies of the } \textit{gene} \text{ (G + g) in the population}}$$

Single nucleotide polymorphisms (SNPs)

SNPs are *genetic markers* used to compare genetic similarities and differences without sequencing the whole genome.



In genome-wide association studies, computers are used to analyse SNPs across genomes, to *compare individuals, establish family lineages,* study *evolutionary relatedness* and indicate *disease susceptibility*.

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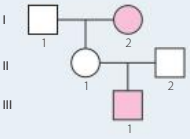
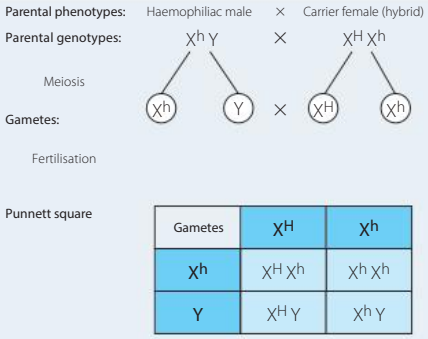
INHERITANCE PATTERNS

Autosomal recessive inheritance



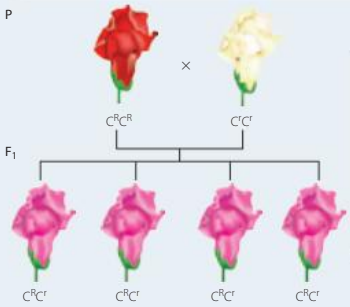
Heterozygotes do not express the trait (recessive). Inheritance is *not related to gender* (autosomal). The trait may *'skip' a generation*. Two parents who don't show the trait produce offspring with the trait.

Sex-linked inheritance



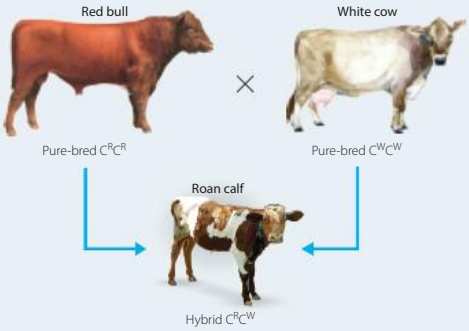
Genes *carried on sex chromosomes* (X and Y) code for traits other than gender. The inheritance pattern differs in male and female offspring.

Incomplete dominant inheritance



When purebreds mate, a *blending* of the trait is expressed in the offspring.

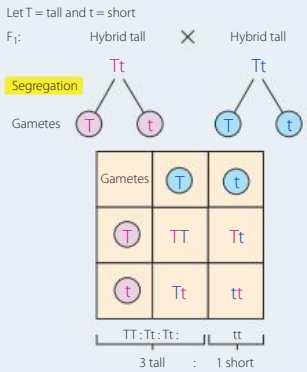
Co-dominant inheritance



When purebreds mate, *both alleles* are expressed in the offspring.

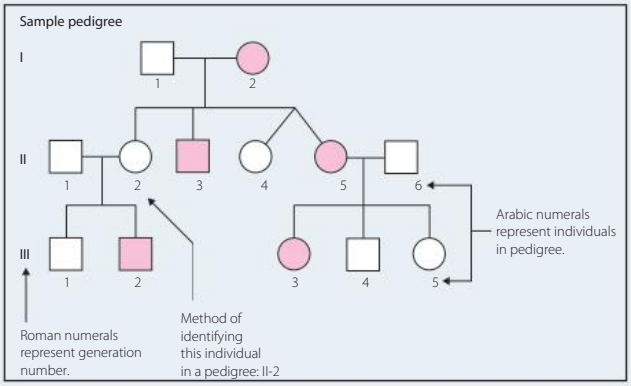
INTERPRETING INHERITANCE PATTERNS

Punnett squares



Punnett squares are a problem-solving technique *used to calculate the probability of a genetic trait being passed on* in a population. It takes into account all possible allele combinations that may arise during fertilisation.

Pedigree charts



Pedigree charts are used *to trace the inheritance of particular traits through several generations* of a family. They use an accepted format of symbols and numbering to compare genetic similarities and differences.

5 CHAPTER REVIEW QUESTIONS

Review quiz

1 Copy and complete the table below to compare mitosis and meiosis. *Note:* If any of the processes occurs in one type of division and not the other, state 'does not occur' under the relevant heading and describe the process (and when it occurs) under the heading of the other type of cell division.

	MITOSIS	MEIOSIS
Cells in which it occurs		
Purpose		
Number of cell divisions to complete the process		
Replication of chromosomes		
Formation of bivalents		
Crossing over		
Alignment at equator		
Halving of chromosome number		
Number of resulting cells		
Genetic content of resulting cells		

2 Explain how meiosis introduces genetic variation into a population.

3 Use labelled diagrams to represent metaphase and anaphase of meiosis I and meiosis II.

4 Draw a diagram to show crossing over of genetic material between a pair of chromosomes. Label two pairs of genes on each chromosome, depicting the alleles as being heterozygous for each gene.

5 Distinguish between the following terms. You may include diagrams and/or examples in your explanation.

- **a** chromosome and chromatid
- **b** autosomal dominant and recessive alleles
- **c** recessive sex-linked allele and recessive autosomal alleles
- **d** codominant alleles and incomplete dominant alleles
- **e** a trait determined by multiple alleles and one determined by multiple genes

6 Explain what is meant by the following terms:

- **a** F_1 hybrid
- **b** pure-breeding homozygote.

7 A black mouse mates with a white mouse and all the offspring are black.

- **a** Explain why there are no brown offspring.
- **b** If the black offspring are interbred, what are the expected genotypic and phenotypic ratios for their offspring? Use a Punnett square to illustrate your answer.

8 Answer the following questions about SNPs.

- **a** What is meant by 'single nucleotide polymorphism'?
- **b** Explain how SNPs arise.
- **c** What is the difference between a SNP and a mutation in a population?
- **d** Why are SNPs useful in genetic studies?

9 Explain what a GWAS is, how it is conducted and the applications of these studies.

10 The ABO blood groups in humans are determined by three alleles at one locus: I^A, I^B and I^i.

- **a** Is it possible for one person to have all three alleles? Explain your answer.
- **b** Which alleles are codominant? Explain what this means.
- **c** Explain why it is not possible to know the genotype of a person who belongs to the blood group A or B unless you investigate the genotypes of their parents and/or offspring.
- **d** What possible gametes will a person with the blood group AB produce? Explain your answer.
- **e** Use a Punnett square to show the phenotypic and genotypic ratios that result for the offspring of a couple with blood groups AB and AO.

11 Draw the standard symbols used in a pedigree to represent:

- **a** parents (both unaffected)
- **b** twins, one unaffected male and one affected female
- **c** the Roman numeral indicating the generation of grandchildren.

12 Look at the following pairs of alleles and identify which genotypes would have a similar phenotype, if the alleles presented by the capital letters are dominant over the alleles presented by the small letters.

AA Aa aa Bb BB bb

13 A breeder of guinea pigs needs to find out whether one of her brown guinea pigs is heterozygous or homozygous for hair colour. Albino (white) guinea pigs have the genotype aa. Guinea pigs that are not albinos have the genotype AA or Aa. Explain how the breeder might do a test cross to determine the genotype of her brown guinea pig. (See page 165 for test crosses.)

14 If you were carrying out breeding experiments with a group of organisms that are heterozygous for a particular gene that has one dominant and one recessive allele:

- **a** How many different phenotypes of offspring would there be? Identify them and give their expected ratios.
- **b** How many different genotypes of offspring would there be? Identify them and give their expected ratios.

15 Explain why it is the male parent in humans who determines the gender of offspring. Use a Punnett square to support your explanation.

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16 A homozygous pea plant with purple flowers is crossed with a homozygous pea plant with white flowers. All the offspring are purple.

- **a** Which colour is dominant? Justify your answer.
- **b** Use a Punnett square to show the possible genotypes that would result from this cross.
- **c** Calculate the ratios of phenotypes and genotypes of the offspring.

17 Explain why each of the following statements is false.

- **a** Offspring resulting from self-fertilisation are genetically identical.
- **b** In a monohybrid cross Bb × Bb, there is a 25% chance of a child being bb. If the first child is bb, there is less of a chance that the second child will be bb.

18 What type of inheritance pattern is shown in Figure 5.33? Justify your answer. Use a Punnett square to show the genetic cross with genotypes of each flower.

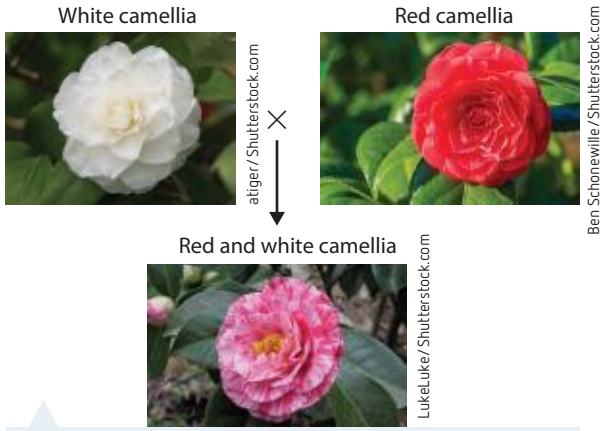


FIGURE 5.33 Genetic cross with camellia flowers

19 Refer to the pedigree in Figure 5.34 to answer the questions below.

- **a** Name the trait being investigated in the pedigree.
- **b** What was the phenotypic ratio of offspring born in the second generation to the grandparents I1 and I2?
- **c** Write a test cross that could be carried out to show whether the trait inherited by the sister with a widow's peak is dominant or recessive. Explain your reasoning.

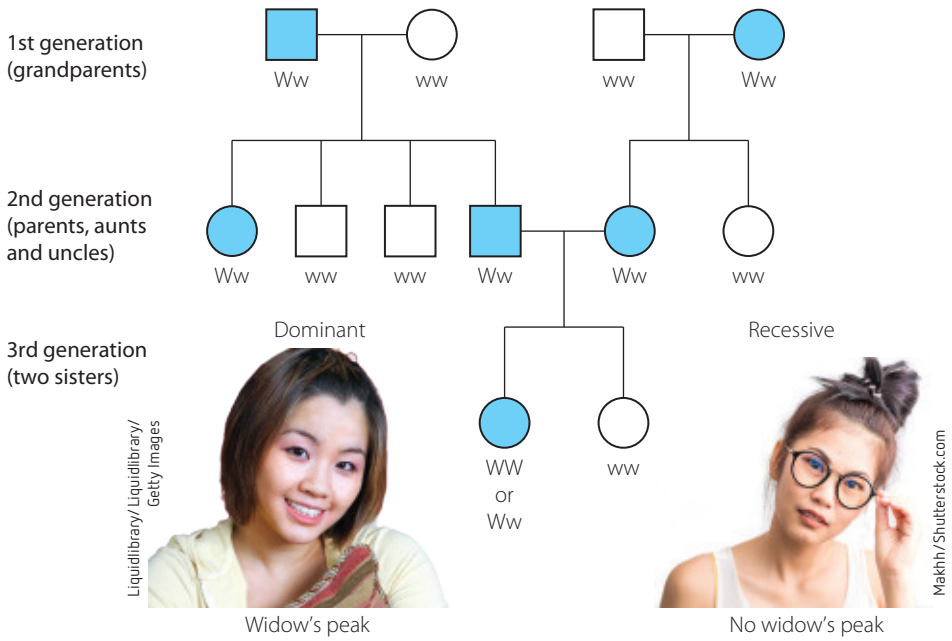
20 Explain how population geneticists measure allele frequency in a population. Include the formula that they use.

21 What are the advantages of using SNPs rather than whole-genome DNA sequencing to identify variations in individuals?

22 Discuss the limitations of SNP frequency data.

23 Select a human genetic disease for which a GWAS has been conducted and explain why, despite being able to identify disease susceptibility in particular individuals, the challenge to establish causality still remains.

Exam preparation



Source: Adapted from pbworks.com

FIGURE 5.34 Pedigree for Question 19

6 Inheritance patterns in a population

INQUIRY QUESTION

Can population genetic patterns be predicted with any accuracy?

Students:

- investigate the use of technologies to determine inheritance patterns in a population using, for example: (ACSBL064), (ACSBL085) ICT
  - DNA sequencing and profiling (ACSBL086) EU
- investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example: (ACSBL064, ACSBL073) AAEA, CCT, IU, N
  - the use of population genetics data in conservation management S
  - population genetics studies used to determine the inheritance of a disease or disorder CCT, ICT, N
  - population genetics relating to human evolution IU

science photo/Shutterstock.com

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
6.1 A secondary-source investigation using DNA technology to study guira cuckoo families

6.2 Secondary source investigation of population genetics data and conservation of koala (Phascolarctos cinereus)

6.3 Secondary-source investigation to determine the inheritance patterns of breast and ovarian cancer genes in a population

6.4 A secondary-source investigation into using genetic evidence to map the trail of our ancestors out of Africa

Worksheets
- Inheritance patterns in a population crossword
- Mitochondrial DNA - an introduction
- Identification of the remains of King Richard III using mtDNA analysis
- Human evolution

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

In 2003, an international group of scientists led by the codiscoverer of the structure of DNA, James Watson, sequenced the first human genome. The publicly funded Human Genome Project (HGP) sequenced DNA obtained from a number of individuals, with the resulting genome representing a mosaic rather than the genome of one person. The final genome was published online, enabling free redistribution and scientific use of the data.

In an almost parallel commercial venture, a corporation known as Celera Genomics sequenced the second human genome in 2008.

The results of both projects have given scientists an improved understanding of many aspects of genetics, including genetic disorders, disease diagnosis and predisposition to disease. This knowledge provides scientists with the potential to individualise diagnosis and treatment of diseases. However, it also gives employers and health insurers information that could be used to discriminate against people on the basis of their future health.

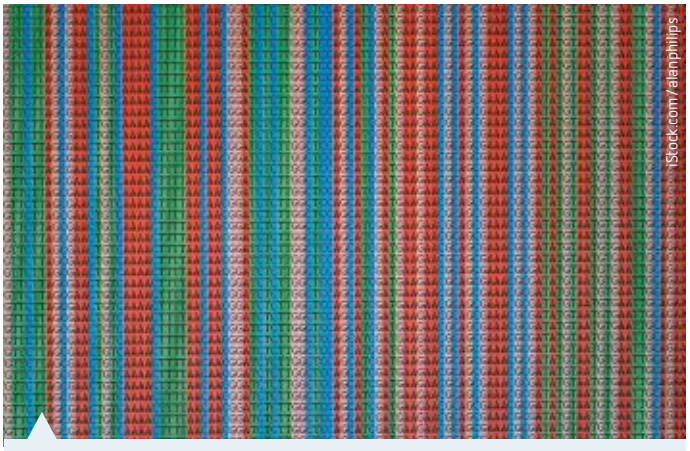

FIGURE 6.1 The Human Genome Project has provided scientists with information about the approximately 3 billion bases that make up approximately 21 000 genes on chromosomes. It has also provided information about approximately 4000 genetic disorders and the locations of potentially faulty genes.

The ramifications of gaining such knowledge are huge. Would you want to know about possible health issues? How do you think this knowledge could be used in the hands of employers and insurance companies?

6.1 DNA technologies

Once the structure of DNA had been unraveled by Watson and Crick, scientists were able to gain more information about how DNA influences the normal functioning of every living thing. They were able to determine the sequence of genes along DNA using techniques such as DNA sequencing and DNA profiling.

In DNA sequencing, the precise order of nucleotides in a sample of DNA is determined. In DNA profiling, an organism's unique DNA profile is determined and represented as a distinct series of bands. Both techniques are described on the following pages.

Worksheet
Inheritance patterns in a population crossword

DNA sequencing

In **DNA sequencing**, the exact nucleotide sequence (the order of the bases A, T, G and C) of a gene on a chromosome is determined. There are several methods of sequencing DNA, including manual methods such as the Sanger chain termination method or the Maxam-Gilbert method, or automatically using a DNA sequencer.

The Sanger method

British biochemist Fred Sanger and his team in the HGP first used the Sanger method to determine DNA sequences. The method is also known as *dideoxy DNA (ddDNA) sequencing*, and when fully automated allows the sequencing of approximately 1000 bases per second.

The first step in this method is to isolate the DNA from the cells of the organism. This is followed by the sequencing reactions (Fig. 6.2). The fragments of DNA produced are then sorted by length using a process called capillary electrophoresis, and the results are analysed by a computer.

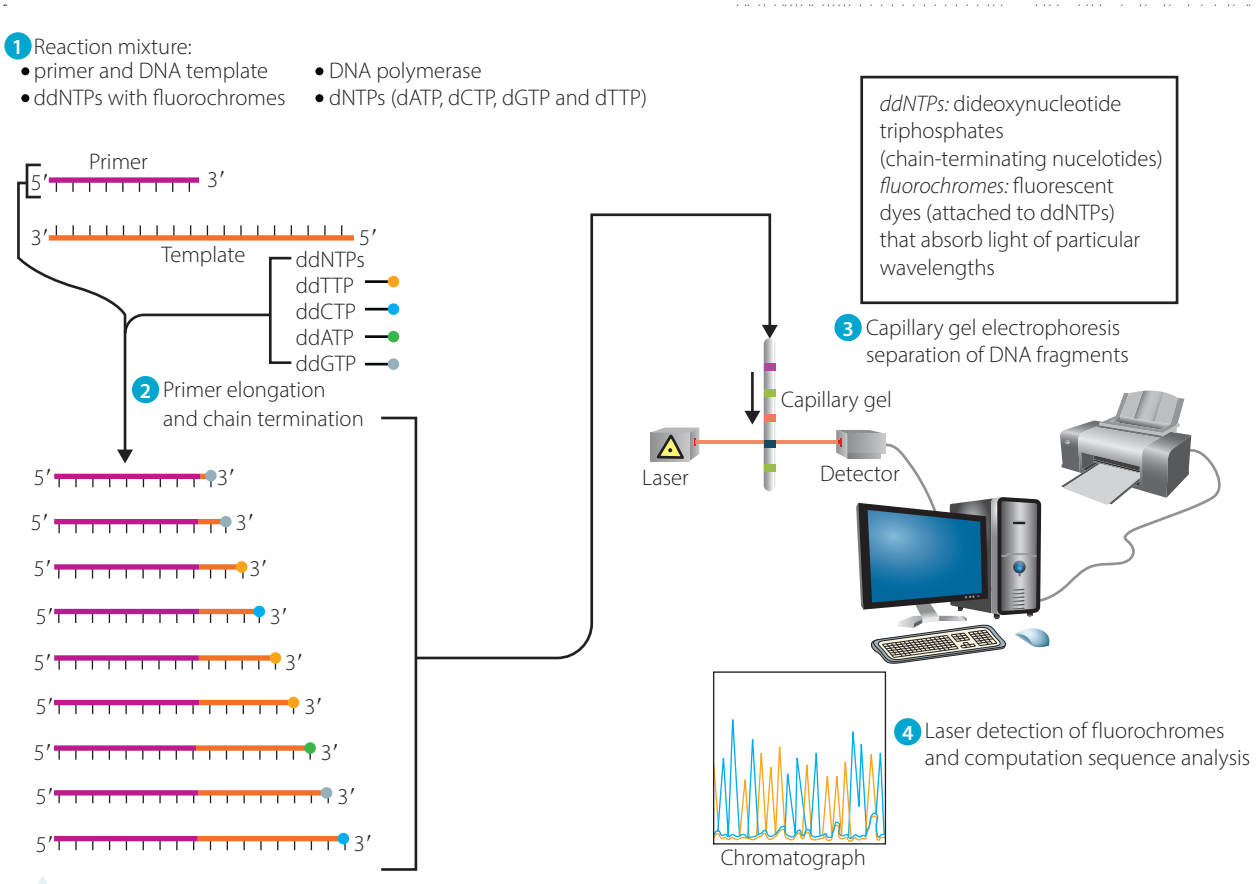

FIGURE 6.2 Steps in the Sanger method of sequencing DNA

Once the DNA has been isolated and replicated, usually by the polymerase chain reaction (PCR) method (page 197), the sequencing reactions are begun. These are done in a series of steps.

1. The double-stranded DNA is separated into single strands by heating.
2. A small piece of DNA called a **primer** binds to the start of the single strand of DNA.
3. DNA polymerase uses this single DNA strand as a template to build the complementary strand of DNA using free nucleotides.

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4 Within the reaction mixture are chain-terminating nucleotides known as dideoxy nucleotides. They are called 'chain terminating' because, after they have attached to their complementary base on the template strand, they prevent any further nucleotides from attaching. There are four types of dideoxy nucleotides, one for each of the bases A, T, C and G: dd**A**TP, dd**T**TP, dd**C**TP, dd**G**TP.

5 Each of the four types of terminator nucleotides is labelled with a different fluorescent dye and will randomly combine with the complementary base on the strands of template DNA. As soon as they become part of the chain, no further nucleotides can attach and the chain is terminated. Chain termination occurs at different positions along the DNA, leading to many different lengths of dye-labelled DNA. The colour of the fluorescent dye is determined by which nucleotide is at the end of the chain. All fragments ending with a particular base have the same colour.

6 This process continues until every position on the template strand has been identified with a chain-terminating nucleotide.

7 The dye-labelled DNA strands are placed into a tiny capillary tube containing a gel, and an electric current is used to pull the strands through the gel in the capillary tube. Shorter strands move through the gel quickly and emerge before the longer strands, which make slower progress through the gel.

8 When the strands emerge, they pass through a laser beam, which causes them to glow at a particular wavelength detected by a photocell and fed to a computer. The wavelength at which they fluoresce depends on the particular base nucleotide. The order in which the complementary nucleotides pass through the laser beam allows the sequence of the bases on the template strand of DNA to be determined.

9 The computer analyses the colours and displays a chromatogram of the sequence of bases in the original DNA sample. In the example shown in Figure 6.3, the sequence of bases on the original DNA strand would be TGAGTCTACGA (complementary to the sequence obtained by reading, in Figure 6.3, the last base on each row, from the bottom to the top).

Weblink
Sanger method
Watch the video DNA Sequencing.

Weblink
Sanger process of DNA sequencing
View this interactive animation and answer the questions.

Weblink
Gel electrophoresis

ACGTACGTACTCAGATGCT
ACGTACGTACTCAGATGC
ACGTACGTACTCAGATG
ACGTACGTACTCAGAT
ACGTACGTACTCAGA
ACGTACGTACTCAG
ACGTACGTACTCA
ACGTACGTACTC
ACGTACGTACT
ACGTACGTAC
ACGTACGTA

FIGURE 6.3 Strands of DNA of different lengths, each beginning with a primer (pink). Nucleotides (black) are added until a fluorescent chain-terminating nucleotide (coloured) is added. The order of the complementary bases on the different lengths of dye-labelled strands of DNA allows the sequence of bases on the template DNA to be determined.

The Maxam-Gilbert method

The Maxam-Gilbert method of DNA sequencing involves chemical sequencing of the DNA strand, and was developed at about the same time as the Sanger method. It is no longer widely used for DNA sequencing, because of its complex nature and its use of hazardous chemicals. It does still have important applications in the further study of DNA, including the structure of nucleotides and modifications made to DNA.

This method involves chemical reactions that are specific to the two groups of bases: pyrimidines (C and T) and purines (A and G).

The first step in the process is to radioactively label one end of the DNA by adding a radioactive phosphorus atom to the phosphate molecule (Fig. 6.4, page 196).

Chemicals and conditions are then modified to suit a specific base, which results in the base being removed from the ribose sugar it is attached to. The DNA strand is cleaved (cut) at this site. This occurs at each position of that specific base in the DNA strand being sequenced.

The DNA fragments formed then undergo gel electrophoresis, with the shorter strands moving further through the gel than the longer strands.

The DNA strand is then exposed to chemicals and conditions that are specific to each of the other bases and the same procedure is followed. When all patterns from the gel electrophoresis are compared, the sequence of the bases on the DNA strand can then be determined.

For example, a strand of DNA is radioactively labelled at one end and the chemicals and conditions for cytosine (C) are applied to the mixture. This causes the removal of the C base from the ribose sugar it was attached to and the cleavage of the strand where it was removed. The same thing occurs at each of the positions of the cytosine, creating fragments of different lengths. The next reaction is to add chemicals and conditions that remove both the C and T bases and then compare the resultant gel electrophoresis to determine the position of the T bases.

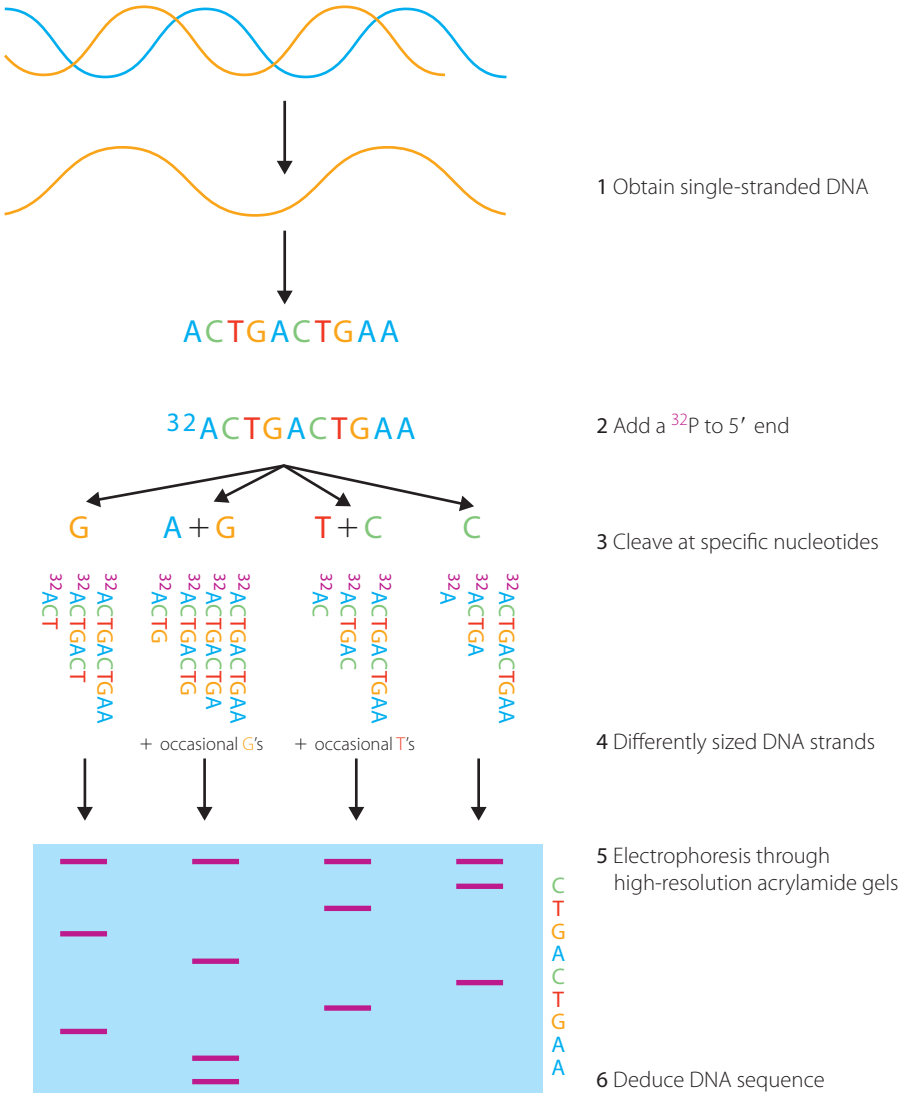

FIGURE 6.4 Steps in the Maxam-Gilbert method of sequencing DNA

FIGURE 6.5 Example of a DNA sequencing device: the portable MinION (produced by Oxford Nanopore) sequences both DNA and RNA and plugs into a computer via a USB.

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Similar reactions are carried out to identify the positions of the G and A bases so that the whole sequence can be determined.

Next-generation technologies

A range of next-generation sequencing technologies are faster, cheaper, use shorter DNA lengths and can sequence many fragments at the same time.

One example is nanopore sequencing. This process involves propelling a DNA molecule with the use of a motor protein through a protein nanopore, a nanometre-sized pore in a membrane that separates two compartments, both containing a buffered potassium chloride solution (Fig. 6.5). The difference in current generated by the aqueous solutions from both chambers when passing through the nanopore depends on the identity of the DNA sequence. This technology allows parallel readings on an individual device, and can control up to 2048 individual nanopores capable of processing 450 base pairs per second.

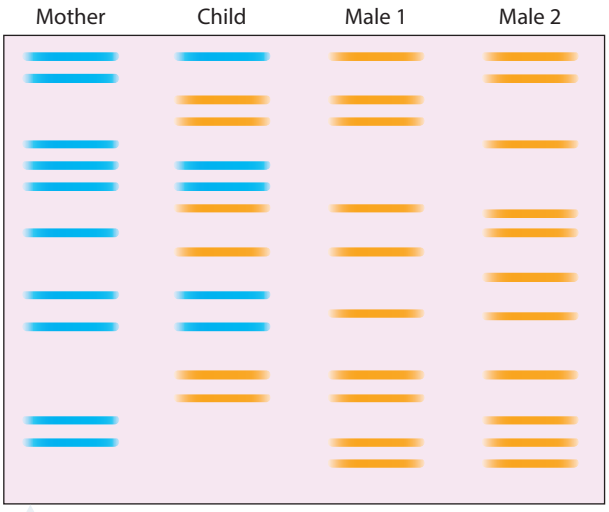
The Human Genome Project cost almost $100 million dollars; in 2018 using this technology it would have cost significantly less.

DNA profiling

DNA profiling, also known as **DNA fingerprint analysis**, is a scientific technique used to identify and compare individuals by characteristics in their DNA. The process was developed by Sir Alec Jeffreys, and has become a useful tool in forensic investigations, paternity testing and other biological applications. The more closely two individuals are related, the more similar their DNA profiles are. The chance of two people who are not identical twins having the same DNA profile is less than one in one billion.

While 99.9% of DNA is common to all humans, there are sections in our DNA called STRs that are unique to each individual. **STRs (short tandem repeats)** are sections of non-coding DNA that are repeated many times over (for example, TATATATATA). The number of repeats at any given location in the non-coding regions of DNA varies between individuals and gives rise to the different DNA profiles. As each individual has two alleles for each STR (one from each parent), the length of a large number of STRs can be used as a basis for identification.

The process of DNA profiling involves isolating the DNA to be profiled. This can come from a variety of nucleated somatic cells such as saliva, blood or cheek cells. The **polymerase chain reaction (PCR)** is used to increase, or amplify, the amount of DNA of the sequence under study. Gel electrophoresis is used to separate the segments according to length. Smaller fragments will have fewer repeats than larger fragments, and so will migrate further on the gel. In Figure 6.6, the mother's DNA is on the left. The child whose paternity is under question is next, and the two potential fathers are on the right (Male 1 and Male 2). Notice that the blue banding of the child matches the blue banding of the mother. The orange banding of the child matches more closely with the orange banding of Male 1 than Male 2.



Source: Adapted from bloodanddna.weebly.com

FIGURE 6.6 Paternity can be established using DNA profiling.

Ethical considerations

Ethical understanding

Once a person's DNA has been analysed, questions must be asked, such as: Who owns the information obtained? Does it belong to the person who supplied the DNA, the company that performed the profile, or another party? Who can access that information?

Consider a situation where the person profiled is carrying the DNA for a disease such as Huntington's chorea. This disease is a progressive brain disorder that causes uncontrolled movements, emotional problems and loss of cognition. It develops as a result of a mutated HTT dominant gene inherited from a

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Can your genes be patented in Australia?

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Can your genes be patented in the US?

parent. The symptoms of this disease do not become apparent until an individual is aged in their thirties or forties. Individuals might not wish to know that they will develop this condition within the next twenty years or so, and children of a known sufferer are faced with the dilemma of whether to be tested. This is in addition to all the other issues faced by people with any inherited condition.

Life insurance companies could access DNA profiling results to determine the risk of insuring a person. If that person is shown to have a higher than normal risk for heart disease or cancer, or will develop a disease such as Huntington's chorea, is it right to charge that person a higher premium? Under existing Australian law, life insurance applicants must disclose the results of any genetic testing if asked to by the insurer. The insurer is then allowed to use these results to discriminate against applicants for life, permanent disability and income protection insurance. This can deter people from undergoing genetic testing or becoming involved in medical research where genetic testing may be involved. Many other countries have protected consumers by restricting or banning the use of genetic testing for insurance purposes.

In order to recoup the immense cost of medical research, some companies have applied to patent gene mutations such as those responsible for BRCA1 and BRCA2. A patent would prohibit all future research and development by another company, and would in effect stifle research. In 2013, a US court ruled that genes cannot be patented; the ruling overturned thousands of US patents on genes.

INVESTIGATION 6.1

A secondary-source investigation using DNA technology to study guira cuckoo families

Cuckoos are medium-sized birds, and there are many different types. The guira cuckoo lays its eggs in communal nests, which have been built by all members of the group, who also share the incubation of the eggs. Female guira cuckoos incubate and raise chicks from eggs that they did not lay.

FIGURE 6.7 The guira cuckoo

One way to determine whether a female guira cuckoo is the biological mother of the chicks she raises is to create a DNA profile of mother and chicks.

AIM

To determine, using DNA profiling, whether guira cuckoo mothers raise their own chicks

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MATERIAL

Ruler

METHOD

1 Consider the DNA profile in Figure 6.8. DNA was obtained by capturing cuckoos, collecting a small blood sample from each, and extracting the DNA. Five STRs have been identified in cuckoos: *Cam*1, *Cam*2, *Cam*3, *Cam*4 and *Cam*5. Using PCR, these five regions were amplified in all adults and chicks of eight families. The PCR products were then separated using gel electrophoresis.

Figure 6.8 shows the resulting gel. Each individual has two alleles for each STR. Each line (band) represents one of the alleles for the STR. Sometimes only one thicker band is observed, if the individual has two identical alleles.

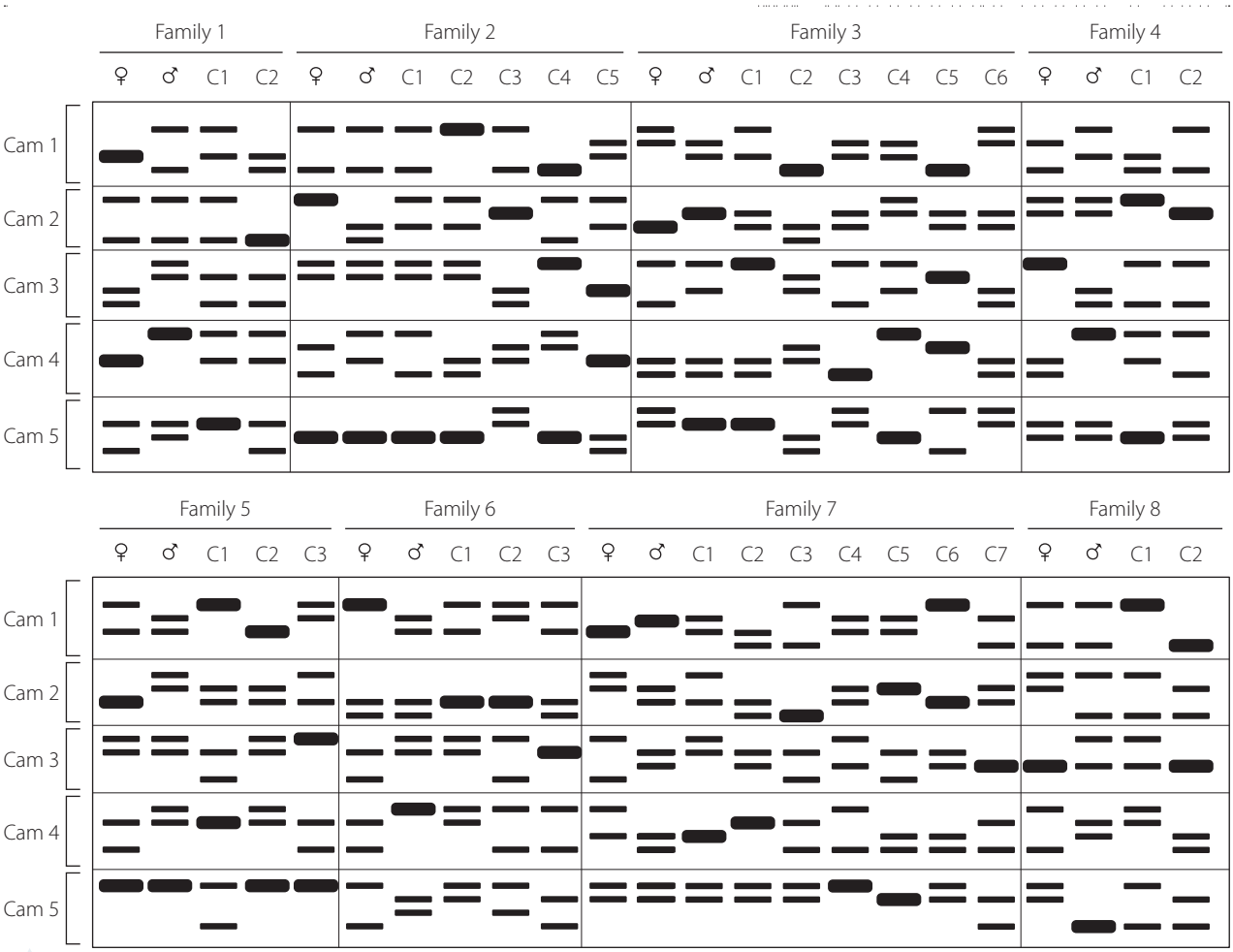

FIGURE 6.8 DNA profiling for the eight cuckoo families that include a social mother (♀), social father (♂) and chicks (C)

2 Compare the profile of the mother of each family and the profile of each chick to determine whether the female could have been the biological mother of the chick or is the social mother. The social mother of the chick is the mother that raises and feeds the chick and does not share genetic similarities.

3 Each chick will have one allele for each STR from its biological mother and one from its biological father. To compare the profile of the mother of each family with the profile of each chick, look at the first STR (*Cam*1) for Family 1. It can be seen that one of the bands for Chick 1 (C1) matches at least one of the bands in the mother. This means that C1 has the same allele for the STR, *Cam*1, as the mother. The same can be seen for C2.

» Continuing for Family 1, when the bands for C1 and C2 are compared with the mother in all of the other STRs (*Cam* 2 – *Cam* 5), it can be seen that both chicks have at least one of their alleles in common with the mother. Therefore, the mother is very likely to be the biological mother of both Chick 1 and Chick 2.

4 Copy Table 6.1 and record in the second column which chicks in each family are the biological chicks.

5 Calculate the proportion of non-family chicks in each family and include these in the third column.

6 Determine whether there is any difference in the proportion of non-family chicks between small and large families.

RESULTS

TABLE 6.1

FAMILY	BIOLOGICAL CHICKS	NON-FAMILY CHICKS
1		
2		
3		
4		
5		
6		
7		
8		

ANALYSIS OF RESULTS

1 Analyse your results to identify (using family numbers and chick numbers) any 'non-family' chicks that have been raised by a social mother cuckoo. Justify your answer, referring to your data.

2 Calculate the maximum proportion of non-family chicks in total over the eight families.

DISCUSSION

1 Explain whether the results show a relationship between family size (large or small cuckoo families) and the likelihood of raising non-family chicks. Use data to justify your answer.

2 How could this practice of social parenting among cuckoos ensure the survival of the species?

3 Discuss the benefits of using DNA profiling in studying populations.

CONCLUSION

Write a conclusion related to the aim of this investigation, to outline your findings.

KEY CONCEPTS

- DNA sequencing determines the exact nucleotide sequence (the order of the bases A, T, G and C) of a gene on a chromosome. DNA sequencing may be conducted using the Sanger method, the Maxam-Gilbert method or next-generation technologies.
- DNA profiling (DNA fingerprint analysis) is a scientific technique used to identify individuals by characteristics such as STRs (short tandem repeats) in their DNA.
- STRs are sections of non-coding DNA that are in the same position on chromosomes for each individual and are repeated many times.
- STRs are unique to each individual because they repeat by different amounts.
- 99.9% of DNA is common to all humans.
- The PCR (polymerase chain reaction) is used to increase the amount of DNA of the sequence.
- Gel electrophoresis is a technique used to separate strands of DNA based on their molecular weight and therefore size. Shorter (lower molecular weight) molecules travel further through the gel than longer (higher molecular weight) molecules.
- Numerous ethical considerations need to be taken into account when using these DNA technologies.

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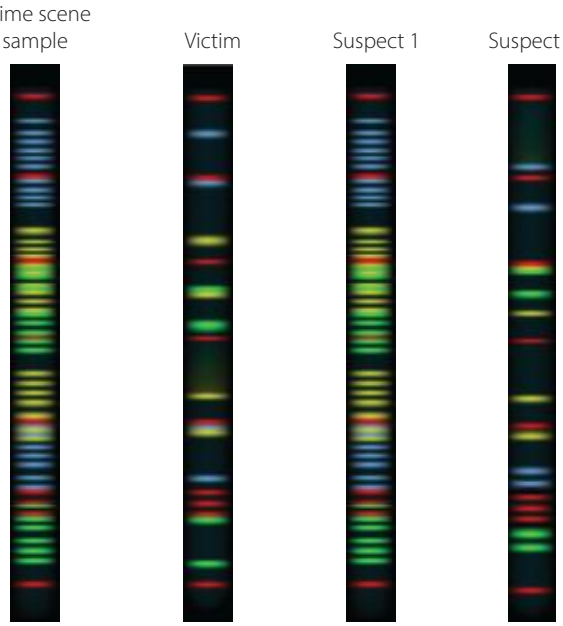
CHECK YOUR UNDERSTANDING 6.1

1 **a** Define 'DNA sequencing'.
   b List three methods of sequencing DNA.
2 Explain how gel electrophoresis separates strands of DNA.
3 Describe how DNA profiling can be used to identify individuals on the basis of their DNA.
4 Give the full name of each of the following abbreviations and explain what each term means.
   a STR
   b PCR
5 A blood-stained sample found at the scene of a jewellery shop break-in was analysed using gel electrophoresis. The police arrested two suspects and also analysed their blood using the same technique. The results are shown in Figure 6.9.
   a The jewellery shop owner also had their blood tested (labelled 'Victim'). Explain why this was necessary.
   b According to the results, which suspect committed the burglary? Explain your choice.
6 Outline three ethical issues associated with use of DNA technologies.



Shutterstock.com / Alila Medical Media

FIGURE 6.9 Analysis of blood from crime scene using gel electrophoresis

6.2 Using large-scale data to study population genetics

With the automation of DNA technologies, scientists have been able to produce large amounts of data that can be saved and analysed by software programs (bioinformatics). This has created many new opportunities for collaboration.

The use of large-scale data sets that are analysed computationally has enabled the scientific community to embark on exciting new research that will have a significant impact on how science is conducted in the future.

Population genetics

Population genetics is the study of genetic variation within a population (Chapter 5), including changes in the frequency of genes and alleles within a population and among populations over time. The alleles of all the genes in a particular population are known as the **gene pool** of the population. Population geneticists study variations in these alleles within the gene pool and how these variations change from one generation to the next. Factors that affect this variation within the gene pool are the size of the population, mutation, natural selection, genetic drift, the diversity of the environment and migration patterns. Genetic differences between species can be used to determine the evolutionary history of populations – those with the most similar gene pools are most closely related.

Refer to Chapter 5 to find out more about Mendel's findings.

Review your understanding of evolution by natural selection from *Biology in Focus Year 11* Chapter 9.

The processes of DNA sequencing and DNA profiling (discussed earlier) and an understanding of heredity allow us to study the frequency of alleles in a population over several generations.

Population genetics depends on the findings of two of the great names in biology. Gregor Mendel showed that each parent provides one copy of each allele to the offspring. The expression of these alleles depends on their pattern of inheritance.

Charles Darwin proposed natural selection as the driving force for evolution. Taking Mendel's and Darwin's work together, we can see that some alleles are selected for as they confer greater survival advantage than others. These advantageous alleles are passed on to the next generation and so increase in number.

Population genetics and conservation management

Conservation genetics is a subfield of population genetics. It incorporates an understanding of how genes are inherited in a population. The principal aim is to avoid extinction of species by applying conservation methods that ensure the maintenance of biodiversity. It combines the principals of applied ecology with an understanding of evolutionary biology.

Traditional methods used by conservation biologists to gather information about endangered species include field observation, sampling and statistical analysis, focusing on the **distribution** and **abundance** of specific organisms. (See *Biology in Focus Year 11*, Chapter 7.)

Conservation genetics relies on gathering genetic data, for biodiversity conservation and to make informed decisions about protecting populations that are endangered or nearing extinction. It is a useful tool for scientists to use when determining current and future strategies for the conservation of populations. The numerous types of DNA analysis, including next-generation analysis methods, use many types of genetic tools including SNPs, GWAS and **haplotypes**, and have been instrumental in determining not only kinship lineages but also in improving our understanding of microevolution, including selection and mutation. These methods also enable scientists to identify segments of the genome that are essential for the organism's adaptation to the environment. They can determine relationships and identify individuals that could be reintroduced into a population for recovery. Any deleterious alleles for brain function, immunity, metabolism and other necessary functions can be detected, as can any mutations that may enhance these functions. These types of information are essential in identifying conservation strategies to increase the chance of saving endangered species and maintaining biodiversity.

Refer to Chapter 5, page 185 for information about SNPs, haplotypes and GWAS.

AFP/Getty Images

FIGURE 6.10 A complete baby mammoth preserved. Researchers have been able to sequence the genome of two mammoths.

Woolly mammoth extinction

One aim of studying population genetics and conservation genetics is to use the past to make accurate predictions about possible future extinction events. Conservation geneticists are hoping that by studying past events they can use the data to develop models that can assist in the conservation of endangered species.

It is possible to study the effects of inbreeding in extinct species. In 2015, scientists conducted a complete DNA sequence to compare two historical samples of the woolly mammoth (Fig. 6.10), to determine whether there was a reduction in genetic diversity due to inbreeding in the population that had been isolated (on Wrangel island). One sample was from a 45 000-year-old specimen that had lived on the Siberian mainland and the other was from a 4300-year-old mammoth bone found on Wrangel Island, in the Arctic Ocean (Fig. 6.11).

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Rainer Lesniewski / Alamy Stock Photo

FIGURE 6.11
Wrangel Island, location of the mammoths that outlived Siberian mammoths by 1500 years. Siberian mammoths died off due to climate change and human hunters.

Genetic sequencing indicated that the island mammoths had a series of major detrimental mutations, including those that affected olfactory processes and some that reduced the number and variety of the mammoths' urinary proteins. These would have greatly reduced their ability to mark and recognise territory, hunt and mate. Another two mutations were found in the gene known as FOXQ1, which affects the mammoths' hair structure. Mutations to this gene would have given the Wrangel Island mammoths a translucent, cream-coloured, satiny coat, which would have reduced its insulating properties, an important feature in ice age climates, where insulation is of paramount importance for survival. It appears that isolation on Wrangel Island and the resultant inbreeding and loss of genetic diversity made these mammoths more susceptible to disease and reduced their ability to survive in their environment. They were said to have suffered a 'genomic meltdown'. Their numbers fell from tens of thousands to around 1000.

The information gained in the mammoth study, about the effects of isolation leading to a reduction in genetic diversity and ultimately extinction, is an example of how population genetics and conservation genetics can be useful tools in trying to conserve modern-day populations that are dwindling in numbers.

Weblink
Extinction of Tasmanian tigers

Using population genetics to study modern koala populations

The koala (*Phascolarctos cinereus*) is a tree-dwelling, medium-sized marsupial with a sturdy body, large rounded ears, grey-coloured fur and sharp claws. Koalas are found throughout eastern Australia, from north Queensland to South Australia. Males are larger than females and a gradient exists for size, where southern koalas are bigger and heavier than those living in Queensland. Their general numbers in the north are declining due to habitat destruction and fragmentation, while the southern numbers are healthy. During the last ice age (about 20 000 years ago), two biogeographic barriers, the Brisbane River Valley and the Clarence River Valley, separated the koala populations.

This impediment to movement disappeared at the end of the ice age, although habitat fragmentation has now restricted the gene flow between populations to some extent.

Koalas are one species, even though historically there has been discussion about a subspecies due to phenotypic differences between geographically distant groups. The results of a very recent large study confirmed that they are one species.

Threats to the koala population include the fur trade in the past (late 19th and early 20th centuries), as well as current habitat clearance. Deaths due to predation, vehicles, diseases such as chlamydia (which affects fertility), bushfires and drought have also reduced their overall numbers.

Often the control of koala numbers is left to the government of the local area, but the results of a recent major study indicate that the overall distribution, both past and present, needs to be taken into account in the management of koala numbers.

The historical and current distribution of koalas is shown in Figure 6.12.

A number of studies have been conducted to investigate the population genetics of koalas (Fig. 6.13), but most of these have been focused at a local level. The recent study referred to above has gathered the largest amount of genetic data ever collected and covers all koala habitats. It also includes information from a range of time periods (1870s to 2015). This study has generated large-scale data sets, which can be analysed. (See Investigation 6.2.)

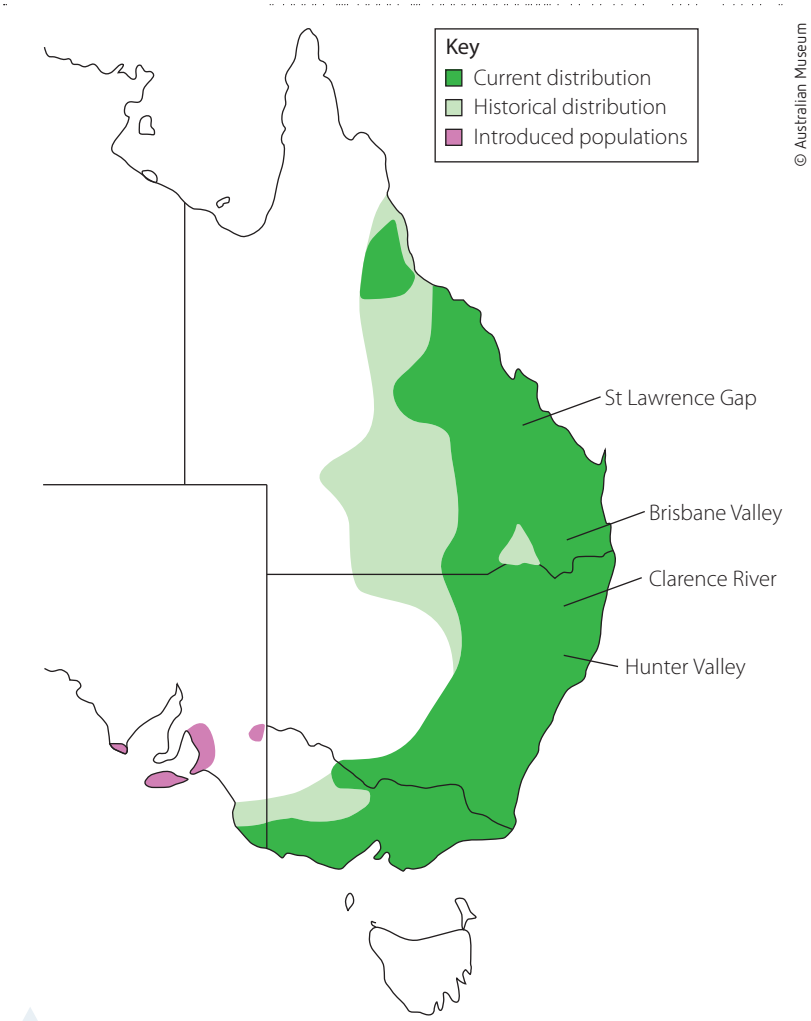

FIGURE 6.12 The current and past distribution of koalas, with four perceived biogeographical barriers labelled. These barriers are thought to have separated koala populations.

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Tissue samples were obtained from 662 wild koalas in collaboration with researchers, consultants, Port Macquarie Koala Hospital and Australia Zoo Wildlife Hospital, and from the Australian Museum Tissue Collection. These samples for DNA analysis were collected from ear punches, to investigate genetic variation in koala populations.


Newspix/Patrick Hamilton

FIGURE 6.13
Testing koalas in order to determine genetic variation in populations

INVESTIGATION 6.2

Secondary source investigation of population genetics data and conservation of koala (*Phascolarctos cinereus*)

Worksheet
Mitochondrial DNA - an introduction

Sustainability

Critical and creative thinking

Numeracy

BACKGROUND

Tissue samples were obtained from 662 wild koalas in collaboration with researchers, consultants, Port Macquarie Koala Hospital and Australia Zoo Wildlife Hospital, and from the Australian Museum Tissue Collection. The mitochondrial DNA control region (mtDNA *CR*) of each tissue sample was analysed by next-generation methods (page 197) to determine the base sequence in this non-coding section of mtDNA. This was used to identify the different haplotypes in the koala tissues being tested. Existing published koala haplotypes that were available in the GenBank databank were also accessed. A *haplotype* is a group of SNP markers, also described as a group of genes that are inherited together on a particular chromosome from the one parent. In the koala study, the genes are inherited down the mother's lineage as mtDNA. Two or more organisms that have the same haplotype will have the same genetic information in that particular position of the chromosome being tested (in this case the *CR* of the mtDNA), indicating that they are closely related to each other. The haplotypes can be used to compare individuals of different populations in order to determine which populations are most closely related each other, and as a result to determine their recent evolutionary history.

A **haplotype network** is a two-dimensional summary of genetic diversity within a group and may be interpreted together with a map of evolutionary and geographical history.

Sometimes these networks look very complicated, so when interpreting a haplotype network you need to remember that:

- Each circle represents a unique haplotype.
- The size of the circle is proportional to the number of individuals sampled that belong to that haplotype.

Haplotypes are groups of genes inherited together on a chromosome from the same parent.

A haplotype network is a summary diagram showing genetic diversity within a group of organisms.

- The lines connect each haplotype to its most similar relative.
- The number of bars on the lines represents the mutational steps between the haplotypes. The more bars, the greater the difference in the sequence of bases between the haplotypes.

A simplified haplotype network is shown in Figure 6.14. This diagram shows three haplotypes in the species being tested, ranging in size from the largest, with 8 individuals sharing a haplotype, to the smallest, with 2 individuals sharing a haplotype. The circle with 8 and the circle with 4 are the most closely related. The bars on the line between 2 and 4 indicate that there are two mutational steps between groups 2 and 4 and, therefore, genetic divergence.

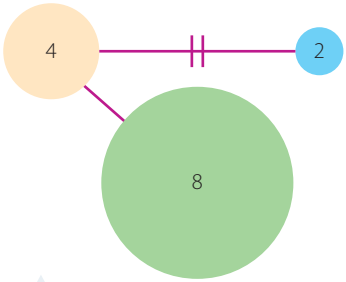


FIGURE 6.14 A simple haplotype network diagram

AIMS

- To investigate the degree of diversity in koala populations
- To interpret genetic data and diagrams and infer trends and patterns

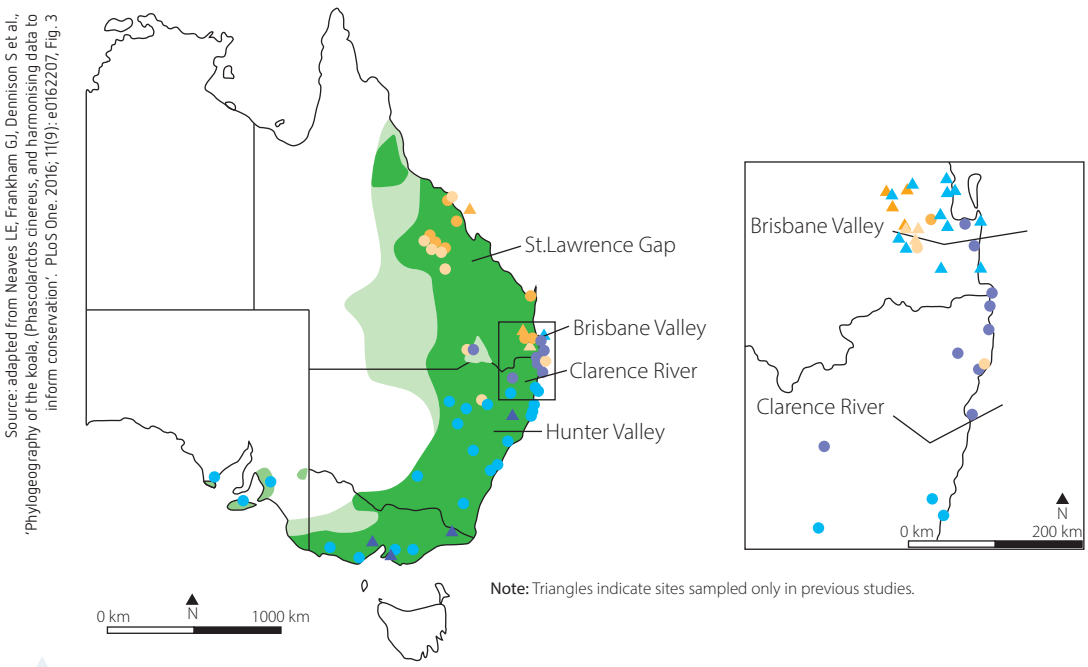
HYPOTHESIS

Propose a hypothesis regarding the genetic diversity of koala populations.

DATA

When the mtDNA *CR* was analysed, 53 unique haplotypes were found across the four regions indicated on the map in Figure 6.15. In Figure 6.16, the four lineages are indicated by the coloured circles:

- orange circles = northern group 1 lineage
- pink circles = northern group 2 lineage
- purple circles = central lineage
- blue circles = southern lineage.



Source: adapted from Neaves LE, Frankham GJ, Dennison S et al., 'Phylogeography of the koala, (Phascolarctos cinereus, and harmonising data to inform conservation'. PLoS One. 2016; 11(9): e0162207, Fig. 3

Note: Triangles indicate sites sampled only in previous studies.

FIGURE 6.15 Geographical distribution of the koala, showing the locations of the sampling of mtDNA *CR* and the four lineages

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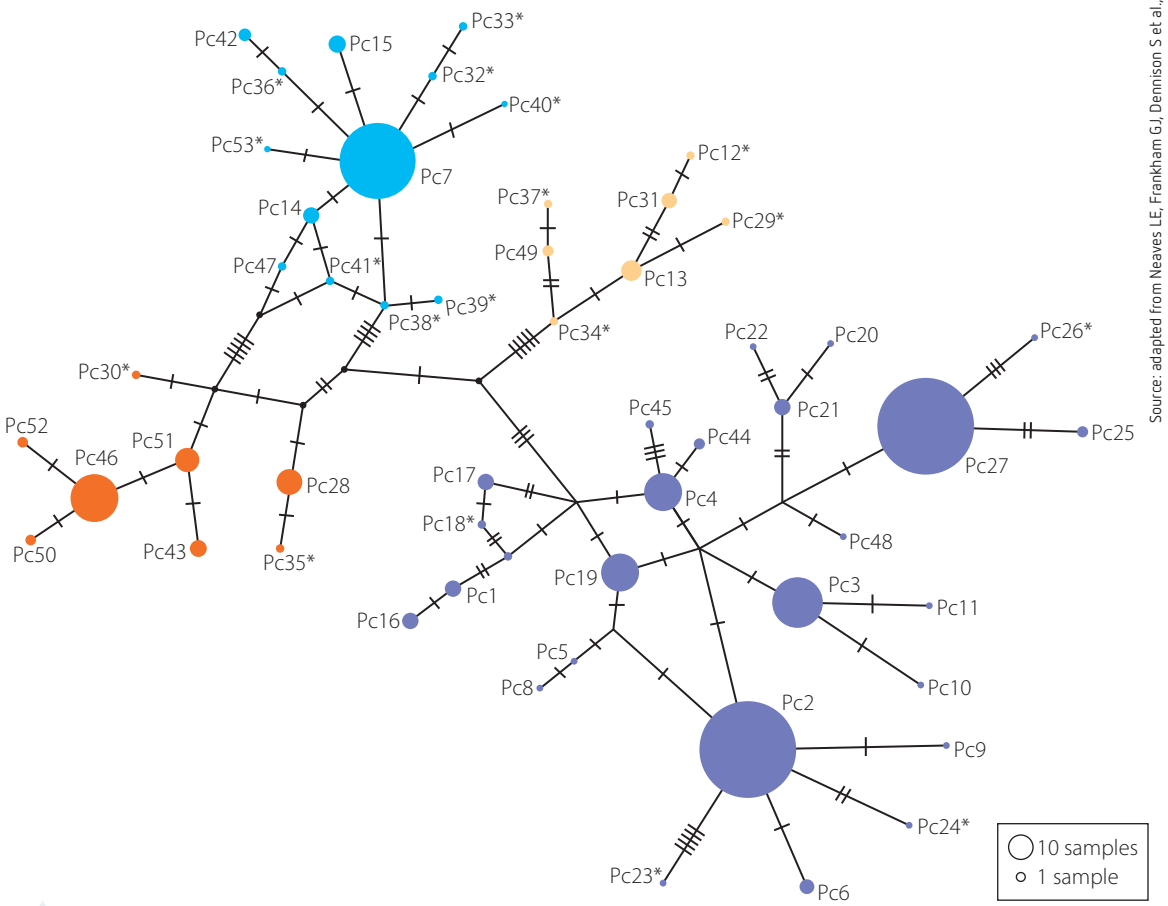
In the haplotype network shown in Figure 6.16, each haplotype is identified by the letters Pc and a number. The size of each circle is proportional to the number of individuals in the group.



Source: adapted from Neaves LE, Frankham GJ, Dennison S et al., 'Phylogeography of the koala, (Phascolarctos cinereus, and harmonising data to inform conservation'. PLoS One. 2016; 11(9): e0116220,. Fig. 4

FIGURE 6.16 Haplotype network diagram for koala mitochondrial DNA *CR*. * means that an individual koala makes up the haplotype (and was recorded from GenBank and not detected in this study).

The data in Table 6.4 reflects diversity in mitochondrial DNA of koalas that were sampled across a range of koala habitats, numbered as locations 1–20 in both Table 6.4 and on the map in Figure 6.17. The table shows the location, sample size, number of unique haplotypes at each location and the haplotype diversity at each location. The higher the value, the greater the haplotype diversity.

Weblink
Koala phylogeography

TABLE 6.4 Mitochondrial DNA *CR* diversity

LOCATION KEY	LOCATION	SAMPLE SIZE	HAPLOTYPES (*n*)	HAPLOTYPE DIVERSITY BASED ON mtDNA
1	Whitsunday Qld	8	4	0.75
2	Blair Athol Qld	10	2	0.20
3	Clermont Qld	38	5	0.37
4	Maryborough Qld	11	1	–
5	Redlands Qld	7	2	0.46
6	Coomera Qld	21	1	–
7	Tyagarah NSW	17	1	–
8	Ballina NSW	37	2	0.074
9	Iluka NSW	7	1	–

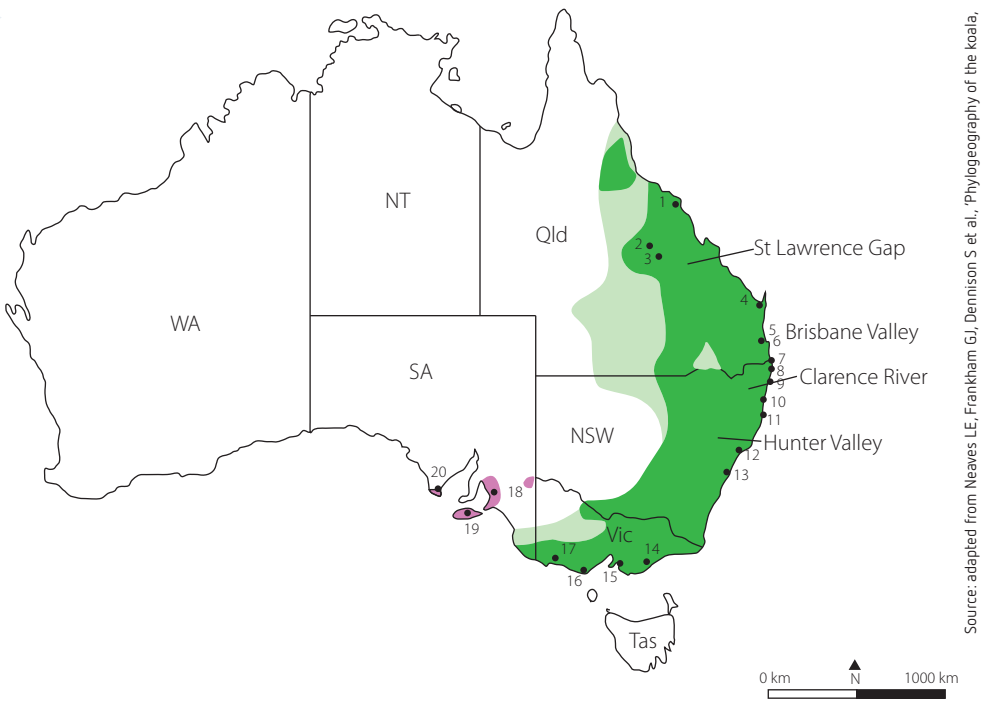
LOCATION KEY	LOCATION	SAMPLE SIZE	HAPLOTYPES (*n*)	HAPLOTYPE DIVERSITY BASED ON mtDNA
10	Pine Creek NSW	50	1	–
11	Port Macquarie NSW	142	3	0.45
12	Maitland NSW	7	1	–
13	Campbelltown NSW	24	4	0.663
14	East Gippsland Vic	19	3	0.119
15	French Island Vic	33	1	–
16	Cape Otway Vic	19	1	–
17	Bessiebelle Vic	14	2	–
18	Mt Lofty Ranges SA	33	6	0.662
19	Eyre Peninsula SA	19	1	0.61
20	Kangaroo Island SA	26	3	0.227
	Overall	662	36	0.84

(Source: adapted from Neaves LE, Frankham GJ, Dennison S et al., 'Phylogeography of the koala, (Phascolarctos cinereus), and harmonising data to inform conservation', PLoS One 2016; 11(9): e0162207, Table 1)

Table 6.5 compares the sequences of DNA in the mtDNA *CR* of koalas distributed across localities 1–20. Numbers 1–20 in Table 6.4, Figure 6.17 and Table 6.5 all denote the same localities. In Table 6.5, koalas from different localities are compared on a pairwise basis. That is, koalas from two regions in different localities are compared to demonstrate genetic diversity. The measurement range is 0–1, where zero indicates that there is no significant diffference, while 1 suggests that koalas are the most different in terms of that haplotype..

Geographically close areas – for example, Coomera (location 6 across the top horizontal row) and Ballina (location 8 down the vertical row) – have a score of 0 (shaded yellow in Table 6.5) indicating no significant genetic difference. Areas with a score of 1 – for example, Pine Creek (site 10) and Iluka (Site 9) – are genetically divergent.

FIGURE 6.17
Map showing sampling sites 1–20 as detailed in Table 6.4



Source: adapted from Neaves LE, Frankham GJ, Dennison S et al., 'Phylogeography of the koala, (Phascolarctos cinereus, and harmonising data to inform conservation'. PLoS One 2016; 11(9): e0162207, Fig. 1

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TABLE 6.5 Pairwise comparison of the sequence of mtDNA *CR* between different localities. Numbers range from 0 (no significant difference) to 1 (most different).

LOCATION	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20
1																				
2	.09																			
3	.21	0																		
4	.57	.78	.75																	
5	.72	.84	.87	.94																
6	.87	.94	.91	1.0	.66															
7	.84	.93	.90	1.0	.62	0														
8	.81	.89	.88	.93	.39	0	0													
9	.75	.88	.88	1.0	.44	0	0	0												
10	.92	.96	.94	1.0	.98	1.0	1.0	.97	1.0											
11	.89	.92	.91	.92	.93	.94	.94	.93	.93	.20										
12	.74	.88	.88	1.0	.94	1.0	1.0	.93	1.0	1.0	.63									
13	.80	.88	.88	.92	.91	.95	.95	.92	.93	.75	.58	.55								
14	.89	.94	.92	.98	.97	.99	.99	.96	.99	.92	.60	.87	.19							
15	.86	.94	.91	1.0	.97	1.0	1.0	.96	1.0	1.0	1.0	.59	1.0	0						
16	.85	.93	.91	1.0	.97	1.0	1.0	.96	1.0	0	.16	1.0	.65	.09	1.0					
17	.90	.95	.93	.99	.98	1.0	1.0	.96	1.0	.97	.61	.95	.25	0	0	.96				
18	.78	.85	.86	.89	.88	.92	.90	.90	.65	.58	.46	0	.19	.18	.55	.50	.22			
19	.86	.94	.91	1.0	.97	1.0	1.0	.96	1.0	1.0	.59	1.0	.20	0	0	.10	0	.18		
20	.83	.90	.89	.94	.93	.96	.96	.93	.95	.76	.56	.57	.09	.01	.02	.67	.04	.09	.02	

(Source: adapted from Neaves LE, Frankham GJ, Dennison S et al., 'Phylogeography of the koala, (Phascolarctos cinereus), and harmonising data to inform conservation', PLoS One 2016; 11(9): e0162207, Table 2)

METHOD

State a hypothesis regarding genetic diversity in koala populations across a range of localities.
Use the data sets provided to answer the questions below.

QUESTIONS

1 a Which section of the koala mtDNA was being tested?
  b Was the same section of mtDNA tested in all koalas?
2 Is this region coding or non-coding?
3 Research and briefly describe the purpose of this region of mtDNA.
4 Refer to Figure 6.16.
  a Determine which lineage has the greatest number of haplotypes.
  b Of the haplotypes Pc2, Pc7 and Pc46, which haplotype:
    i has the highest number of individuals with that haplotype
    ii is part of the central lineage
    iii has the lowest number of individuals with that haplotype?
  c Determine the total number of mutational steps between:
    i Pc19 and Pc27
    ii Pc47 and Pc51.
  d Which of these pairs is more closely related: Pc19 and Pc27, or Pc47 and Pc51?

» 3 Is there is only one mutational step between:
   a Pc3 and Pc10
   b Pc3 and Pc11?
4 Why are Pc10 and Pc11 different haplotypes?
5 Is there greater genetic variation within or between lineages?
6 Refer to Figure 6.17.
   a How many lineages are predominant in the majority of NSW, Victoria and SA (Region 1)?
   b How many lineages are present in the area between far north of NSW up to just above the midpoint of the Queensland coast (Region 2)?
   c Where is most of the central lineage located?
   d In which of the two regions (1 or 2) referred to in questions **a** and **b** would you consider there to be the most genetic diversity? Provide reasons for your answer.
7 Refer to Table 6.4.
   a Which location had the greatest number of haplotypes?
   b Which location has the greatest haplotype diversity?
   c What is the value of the haplotypic diversity for location 20, Kangaroo Island?
   d Identify the lineages for each of the locations you identified in questions **a**, **b** and **c**.
8 Refer to Table 6.5 and Figure 6.17. In each of the following pairs of locations, determine whether the koalas are closely related or genetically divergent. Also state whether or not the locations are close to each other.
   a Region 1: far north NSW – Queensland
      i 1 and 5
      ii 1 and 3
      iii 7 and 4
      iv 10 and 6
   b Region 2: the rest of NSW, Victoria and South Australia
      i 13 and 12
      ii 14 and 13
      iii 17 and 13
      iv 20 and 14
8 In which region, 1 or 2, is the greatest genetic diversity shown within the koala populations? Use the data you have studied to justify your choice.
9 The koala population in locations 19 (Eyre Peninsula) and 20 (Kangaroo Island) were formed by introducing koalas from other locations.

   Use the data in Table 6.5 to predict the likely source(s) of koalas for each of these locations. Justify your answer.
10 Does the data provided support your hypothesis? Explain your answer.

DISCUSSION

1 The greater the genetic diversity in a population, the greater the chance of survival of that population. Which of the lineages in this study shows the least genetic diversity and how would you increase this diversity? Use data that you have studied to support your answer.
2 The data that has been studied here is from one study. How would you improve the validity of the inferences made from this data?

CONCLUSION

Summarise your findings regarding koala genetic diversity in eastern Australia.

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KEY CONCEPTS

- Population genetics is the study of genetic variation within a population, and the changes in the frequency of genes and alleles within a population and among populations over time.
- Mendel's and Darwin's work forms the basis of population genetics, in showing that some alleles are selected as they confer greater survival advantage than others.
- Conservation genetics looks at how genes are inherited in a population, in order to avoid extinction of species by applying conservation methods that ensure the maintenance of biodiversity.
- Genetic analysis using large-scale data including GWAS, SNPs and haplotypes provides detailed information about genetic diversity in a population.
- Haplotype networks map the different haplotypes in populations.
- The extinction of the woolly mammoth on Wrangel Island can in part be attributed to a severe decrease in genetic diversity due to small population numbers.
- The control region of mitochondrial DNA in koalas was analysed to determine the different lineages and genetic diversity in koalas in eastern Australia.
- The southern lineage of koalas demonstrates the lowest genetic diversity compared with the lineages from north-eastern NSW and Queensland, which demonstrate high levels of genetic diversity.

CHECK YOUR UNDERSTANDING 6.2

1. Define 'population genetics' and 'conservation genetics'.
2. How do the findings of Mendel and Darwin influence the study of population genetics?
3. How can DNA analysis be used in population genetics and conservation management? Provide an example.
4. Distinguish between SNPs and haplotypes.
5. State the aim of conservation genetics.

6.3 Using large-scale data to study population genetics and disease

Alkaptonuria (black urine disease) is a disease in which the body cannot process the amino acids phenylalanine and tyrosine, which occur in protein. This disease causes many problems but the first symptom is urine with an unusually dark colour, which turns black if left exposed to air.

In the early part of the 20th century, Archibald Garrod demonstrated that this disease is an inherited recessive disease. He made this deduction by surveying the families of his patients and developing a pedigree of each family history.

With the expansion of technologies such as DNA sequencing, it was subsequently found that alkaptonuria is caused by a mutation in the HGD gene for the enzyme homogentisate 1,2-dioxygenase, located on chromosome 3. Alkaptonuria is an example of a disease that results from a mutation in a single gene in all cells of the body, and is known as a **monogenic** disease.

Scientists currently estimate that there are more than 10 000 monogenic human diseases. Although relatively rare, they affect millions of people worldwide. The global prevalence of all monogenic diseases at birth is approximately 10 in every 1000 births.

With the advent of genetic testing, the diagnosis of these diseases and the probability of any future offspring inheriting these conditions can be determined quickly and accurately.

In humans, the most common genetic differences (**polymorphisms**) in the genome are single base pair differences called **single nucleotide polymorphisms (SNPs)**, pronounced 'snips'. Genetic testing for the presence of the SNPs that are unique to a particular disease can be carried out quickly and relatively easily.

Worksheet
Identification of the remains of King Richard III using mtDNA analysis

FIGURE 6.18 A newborn screening test called the heel prick test provides genetic testing for 25 congenital conditions.

In New South Wales, the newborn screening program provides free genetic tests for all newborns for the SNPs associated with phenylketonuria, congenital hypothyroidism, cystic fibrosis, galactosaemia, fatty acid oxidation, urea cycle disorders and many more **congenital** (present at birth) diseases. Most of these conditions are genetic.

The data collected provides the potential for large-scale genomic analysis of newborns. Such screenings have a significant impact on individuals, through early detection and improved treatment options. They also assist in generating data about the occurrence of specific genetic conditions in the population.

There have been many efforts to create a gene–disease specific mutation database with the increasing amounts of data provided by next-generation gene sequencing technologies. The Human Gene Mutation Database (HGMD) stores comprehensive information about germline mutations associated with human inherited diseases. HGMD does not store information about somatic and mitochondrial mutations, as this is stored in COSMIC (Catalogue of Somatic Mutations in Cancer) and MITOMAP (human mitochondrial genome) databases, respectively.

INVESTIGATION 6.3

Secondary-source investigation to determine the inheritance patterns of breast and ovarian cancer genes in a population

Ethical understanding

Information and communication technology capability

Numeracy

Critical and creative thinking

BACKGROUND INFORMATION

Breast screens involving a mammogram are used to identify abnormalities in breast tissue. One particular type of breast cancer is a 'basal-like' cancer that is particularly aggressive, more difficult to control and harder to treat than most forms of this disease.

Mutations of the BRCA1 and BRCA2 genes put women at a much higher risk of developing this form of breast cancer. The BRCA1 and BRCA2 mutations are inherited and are carried by one woman in 600. The presence of these mutated genes increases the risk of developing this cancer by 85%.

BRCA1 and BRCA 2 are tumour suppressor genes found on chromosome 17 and chromosome 13 respectively (Fig. 6.19). They are believed to be responsible for coding for proteins that repair genes involved in regulating the process of cell division. Mutations of the BRCA1 and BRCA2 genes mean that the genes that control cell division are not repaired, which ultimately results in the uncontrolled cell division associated with cancer.

In an American longitudinal study involving 46 276 women, the genes were sequenced to identify the prevalence of BRCA1 and BRCA2 mutations. Deleterious mutations to the BRCA1 or BRCA2 gene were identified in 12.5% of subjects. The study also looked at the ancestry of the subjects to see if there was a relationship between ethnicity and the prevalence of the mutated genes.

»

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AIM

To use data to interpret trends, patterns and relationships in the inheritance of genes that increase an individual's chance of developing breast or ovarian cancer

METHOD

Complete the following activities and answer the questions regarding the prevalence of BRCA1 and BRCA2 genes in different populations and the risk of developing breast or ovarian cancer.

Go to the weblink to view the study.

PART A

Table 6.6 shows the prevalence of BCRA1/2 mutations in women of different ethnicities, obtained from a large-scale population genetics study.

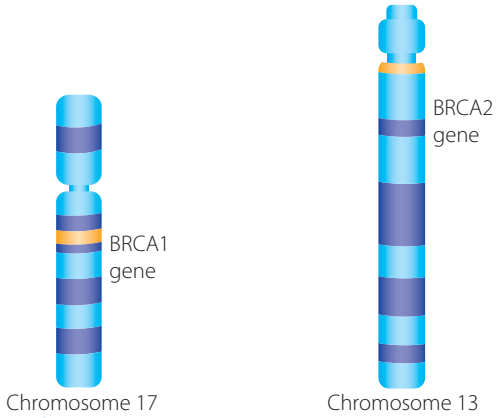


FIGURE 6.19 Chromosome 17 and chromosome 13 showing the location of the BRCA1 and BRCA 2 gene

Weblink
BRAC1 and BRAC 2 mutations information

TABLE 6.6 Prevalence of the BCRA1/2 genes in the study

ETHNICITY	NUMBER OF SUBJECTS	DELETERIOUS MUTATIONS (% OF WOMEN IN STUDY)		
		BRCA1	BRCA2	TOTAL
Western European	36 235	6.9	5.2	12.1
Central European	4 066	8.3	5.3	13.5
Latin American	1 936	9.6	5.4	14.8
African	1 767	10.2	5.7	15.6
Asian	1 183	6.3	6.3	12.7
Native American	597	7.4	5.9	13.2
Middle Eastern	492	6.1	3.3	9.4
Total	46 276	7.2	5.3	12.5

(Source: adapted from Hall MJ, Reid JE, Burbidge LA et al. 2009, 'BRCA1 and BRCA2 mutations in women of different ethnicities undergoing testing for hereditary breast-ovarian cancer', Cancer 115(10): 2222–33, 15 May 2009, Table 2, see http://onlinelibrary.wiley.com/doi/10.1002/cncr.24200/full)

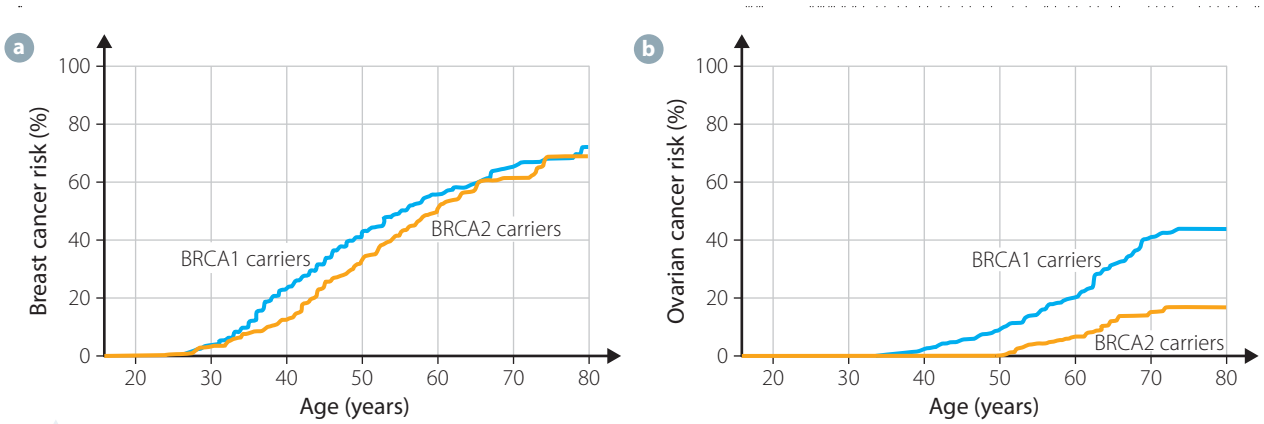
1. Draw an appropriate graph of the data in Table 6.6, indicating the percentage of women in each ethnic group who have the BRCA1 and BRCA2 genes. The *y*-axis should have a suitable scale. The *x*-axis should have two divisions per ethnic group, one for each type of mutation. Include a key for identification of BRCA1 and BRCA2.
2. Using both the graph you have drawn and the information in Table 6.6, answer the following questions.

QUESTIONS

1. Which ethnic group had the highest incidence of:
   - **a** the BRCA1 gene
   - **b** the BRCA2 gene?
2. Outline the general trend exhibited in the relationship between percentages of the BRCA1 gene and the BRCA2 gene for most ethnic groups.
3. Compare the BRCA1 and BRCA2 percentages for women of Western European ethnicities with those of Latin American and African ethnicities.
4. Suggest how the reliability of the results for Native American and Middle Eastern ethnicities could be improved.

PART B

Refer to Figure 6.20 to answer the questions that follow.



Source: adapted from Hall MJ, Reid JE, Burbidge LA et al. 2009, 'BRCA1 and BRCA2 mutations in women of different ethnicities undergoing testing for hereditary breast–ovarian cancer', Cancer 115(10): 2222 – 33, 15 May 2009, Table 2

FIGURE 6.20 Cumulative risk of developing **a** breast cancer and **b** ovarian cancer in carriers of the BRCA1 and BRCA2 gene mutations

QUESTIONS

1 What is the cumulative risk of developing breast cancer at:
   - **a** age 40, in:
     - **i** BRCA1 carriers
     - **ii** BRCA2 carriers
   - **b** age 74, in:
     - **i** BRCA1 carriers
     - **ii** BRCA2 carriers?

2 Outline the general trend shown in the graph for the cumulative risk of developing breast cancer as age increases for BRCA1 carriers.

3 Determine the cumulative risk of developing ovarian cancer at:
   - **a** age 48, in:
     - **i** BRCA1 carriers
     - **ii** BRCA2 carriers
   - **b** age 70, in:
     - **i** BRCA1 carriers
     - **ii** BRCA2 carriers.

4 Outline the general trend exhibited on the graph for the cumulative risk of developing ovarian cancer in BRCA1 carriers and compare this to the trend for BRCA2 carriers.

5 Compare the trends shown for the cumulative risk of developing breast cancer with the cumulative risk of developing ovarian cancer in BRCA1 carriers.

6 Using data from all sources presented in this investigation, determine whether it is possible to predict the chances of developing breast or ovarian cancer.

CONCLUSION

Based on the data studied in this investigation, is it possible to predict the chance of developing breast or ovarian cancer based on the inheritance of the BRCA1 or BRCA2 gene mutation? Justify your answer using data from both sources.

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KEY CONCEPTS

- The prediction of genetic susceptibility to a disease has traditionally been based on family background and pedigree analysis.
- Large-scale screening and DNA analysis can have a significant impact on individuals through early detection and improved treatment options.
- The Human Gene Mutation Database (HGMD) stores comprehensive information about germline mutations associated with human inherited diseases.
- Analysis of large-scale genetic information can be used to predict the inheritance of a disease or disorder.

CHECK YOUR UNDERSTANDING 6.3

1 Before the development of genetic technologies, how was an inherited disease traced within a family?
2 Define these terms: monogenic, polymorphism, single nucleotide polymorphism.
3 What type of data can be obtained from newborn screening tests?
4 Distinguish between the normal and mutated BRCA1 gene.

6.4 Using large-scale data to study population genetics and human evolution

Anthropological genetics is an emerging branch of science that combines components of population genetics, such as DNA analysis, with historical, archaeological and linguistic evidence to determine the pathways of human evolution. It aims to explain the causes of human diversity in our past and present. The evolutionary forces of mutation, natural selection, genetic drift and gene flow are responsible for the patterns of diversity in human populations today. The number of alleles and the frequency of alleles in a population are measures of this diversity. Mathematical population models allow scientists to interpret genetic diversity and predict the genetic patterns associated with human evolution.

Genetic drift and gene flow are discussed in Chapter 7.

Intercultural understanding

Anthropologists have grappled with the question of when modern humans arose as a species. Did they arise in Africa 200 000 years ago, or did they come from a subgroup once they were out of Africa? Studying the human genome rather than fossils alone (Fig. 6.21) has significantly changed our understanding of human evolution.

FIGURE 6.21 Traditional methods of analysis used to study human evolution are based on comparing fossil evidence such as archaic (left) and modern (right) *Homo sapiens* skulls.

Human migration theories

There are two main contesting theories regarding human migration out of Africa: the *Multiregional hypothesis* and the *Replacement hypothesis*.

The **Multiregional hypothesis (MRE)** relies mostly on fossil evidence and suggests that all human populations can be traced back to when *Homo erectus* first left Africa, about 2 million years ago. It is suggested that there was gene flow between neighbouring populations and that once they dispersed into other portions of the old world, they slowly evolved into modern humans (Fig. 6.22).

The **Replacement hypothesis**, also called the Out of Africa or Eve hypothesis, suggests that archaic *Homo sapiens* left Africa. It proposes that a second migration out of Africa happened about 100 000 years ago and that modern humans of African origin conquered archaic groups and replaced them by interbreeding with and out-competing them.

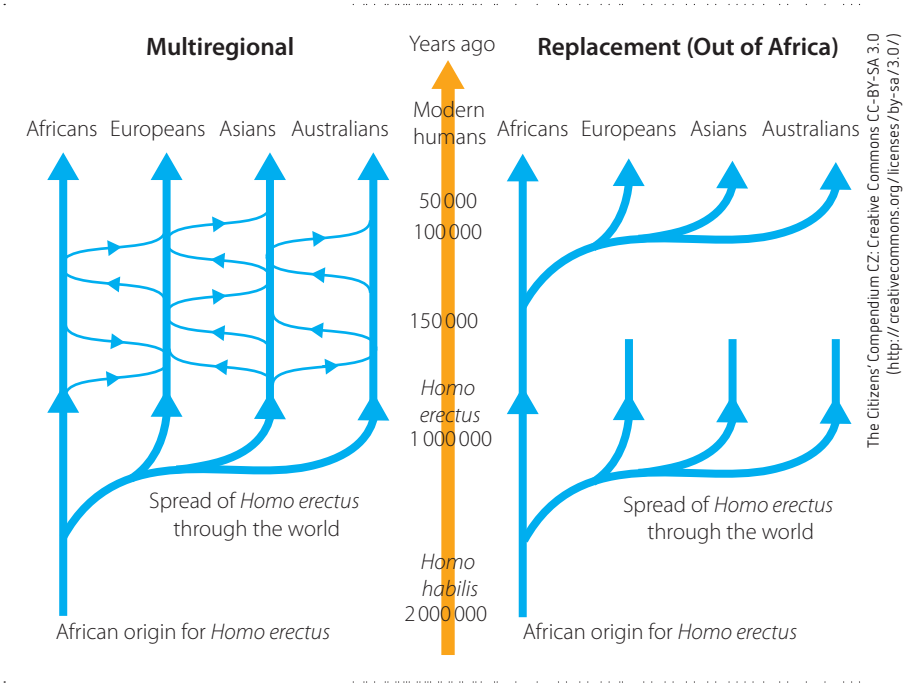

FIGURE 6.22 Comparison of the Multiregional and Replacement hypotheses of human migration out of Africa

Genetic evidence

Modern genetic studies have shown that if the MRE were correct, modern populations would contain ancient alleles scattered in different regions of the world. Researchers sequenced over 18 000 whole human mtDNA genomes from people all over the world.

Mitochondrial DNA was chosen because its pattern of maternal inheritance (hence, named the Eve hypothesis) provided a relatively uninterrupted lineage of descent from ancestral populations. Global human populations were grouped according to the specific mutations in their mtDNA: members of a group that share the same mutations must be descendants of a common ancestor (haplogroups). Using molecular homology, phylogenetic trees were produced from the mtDNA haplogroups.

It was discovered that, among modern humans, most of the variation in mtDNA sequences occurs in African populations (L haplogroups, Fig. 6.23). The mtDNA of Europeans, Asians and the Indigenous peoples of Australia, the Americas and Pacific islands represent just a subset of total human mtDNA diversity (M and N haplogroups, Fig. 6.23). This provides evidence for the Replacement (Out of Africa) hypothesis.

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Molecular clock estimates suggest that diverse populations of modern humans evolved over 200 000 years in Africa, with the haplogroups that migrated out of Africa diverging 70 000 years ago. Superimposing the phylogenetic tree on a map of Africa (Fig. 6.23) strengthens the case for migration. The two surviving mtDNA groups (M and N) that colonised the other continents are most closely related to the African L3 group located north-east of Africa and nearest the Middle East.

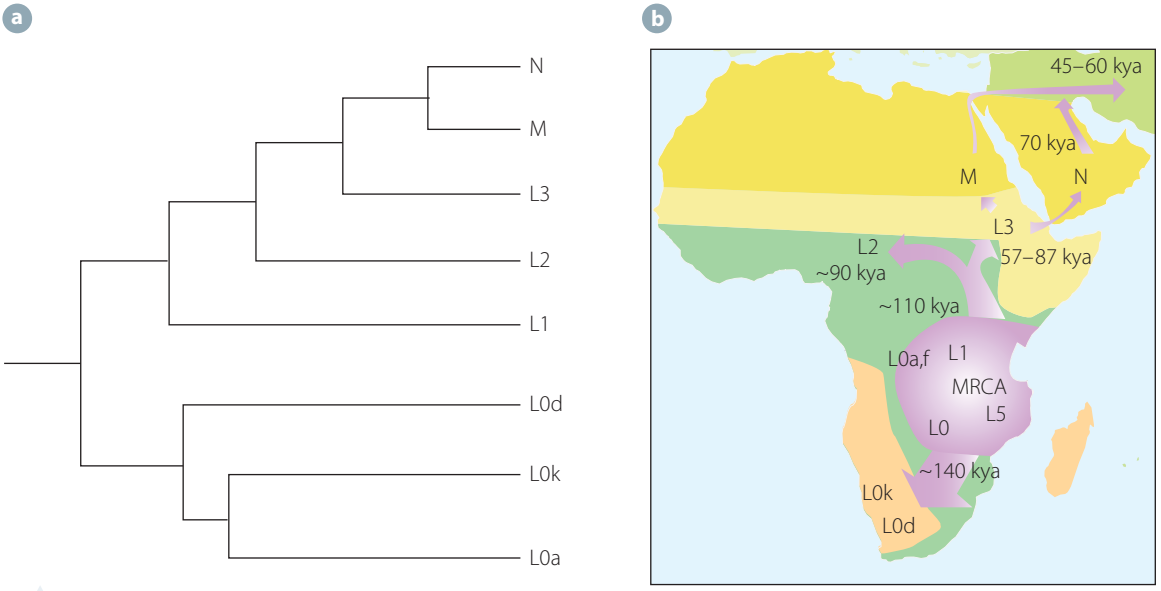

FIGURE 6.23 **a** Phylogenetic tree generated from mtDNA sequences of global human populations. The labels at the tips of the tree represent the haplogroups into which human mtDNA mutations can be classified. **b** Map showing the location of each haplogroup and inferred migration patterns thousands of years ago (kya). All haplogroups originate in Africa and the Middle East. Only the M and N haplogroups are found in indigenous populations throughout the rest of the world. MRCA = most recent common ancestor.

Out of Africa

The evidence suggests that, when our human ancestors first migrated out of Africa, they were not alone. At least two other species of humans (Neanderthal and Denisovans) occupied what is now the Eurasian land mass. Neanderthal (*Homo neanderthalensis*) remains (Fig. 6.24) are found in western, central, eastern and Mediterranean Europe, as well as south-west, central and northern Asia up to the Altai Mountains in Siberia.

Denisovans (Fig. 6.24) were a subspecies of humans. A 41 000-year-old finger bone found in a cave in the Altai Mountains in Siberia is genetically different from both Neanderthals and humans. These caves had been inhabited by both Neanderthals and Denisovans, possibly even overlapping at times.

As our human ancestors migrated through Eurasia, they would have encountered Neanderthals and Denisovans. Genetic evidence suggests that they interbred with Neanderthals. In doing so, a small amount of Neanderthal DNA was introduced into the human genome, and persists today. Scientists have concluded that most Europeans and Asians have 2% Neanderthal DNA. In fact, genetic analysis of the oldest European mummy ('Otzi the iceman', who lived around 3400 BCE) shows it to contain an even higher percentage.

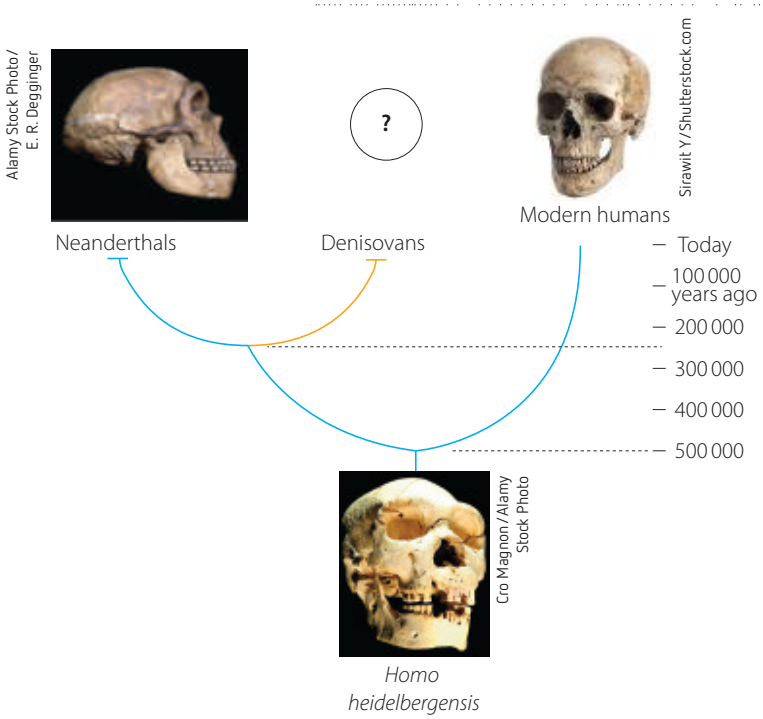

FIGURE 6.24
Neanderthals and Denisovans were closely related. DNA comparisons suggest that the ancestors of modern humans diverged 500 000 years ago.

INVESTIGATION 6.4

A secondary-source investigation into using genetic evidence to map the trail of our ancestors out of Africa

Mitochondrial DNA (mtDNA) is ideal for tracing human evolution because it is inherited only through the maternal line and is not subject to recombination, as nuclear DNA is. There are also 10^{16} molecules of mtDNA within every human, and each mtDNA molecule is usually identical to all the others. The mean rate of mtDNA divergence within humans is 2–4% per million years.

A worldwide survey of mtDNA drawn from 147 people from five geographic regions was conducted and partial results are presented below.

AIM

To use data from secondary-source investigations to determine the route taken by our ancestors when they migrated out of Africa

BACKGROUND

The five geographic regions sampled are:

1. Sub-Saharan Africa ($n = 20$)
2. Asian from China, Vietnam, Laos, the Philippines, Indonesia and Tonga ($n = 34$)
3. Australian Aborigines ($n = 21$)
4. Caucasians from Europe, North Africa and the Middle East ($n = 46$)
5. Aboriginal New Guineans ($n = 26$).

Table 6.7 shows the mtDNA divergence (difference) within and between the five human populations. The shaded diagonal shows divergence between individuals within the population, the cells below the diagonal show the divergence between the two different populations, and the cells above the diagonal have been removed for the purpose of this investigation.

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TABLE 6.7 MtDNA divergence within and between five human populations

POPULATION	SEQUENCE DIVERSION (%)				
	1 AFRICAN	2 ASIAN	3 AUSTRALIAN	4 CAUCASIAN	5 NEW GUINEAN
1 African	0.47				
2 Asian	0.45	0.35			
3 Australian	0.40	0.31	0.25		
4 Caucasian	0.40	0.31	0.27	0.23	
5 New Guinean	0.42	0.34	0.29	0.29	0.25

(Source: Adapted from Cann RL, Stoneking M & Wilson AC 1987, 'Mitochondrial DNA and human evolution', Nature 325: 321-36, Table 1)

QUESTIONS

1 a Which population shows the greatest sequence divergence in mtDNA sequences within the population?
  b What does this mean in terms of the age of this population?
2 a Which population shows the least divergence in mtDNA sequences within the population?
  b What does this mean in terms of the age of this population?
3 Formulate a hypothesis to explain the observations in Questions **1** and **2**.
4 Explain why sub-Saharan Africans do not have any Neanderthal DNA in their genome.
5 Use evidence from Table 6.7, Figure 6.25 and pages 216–18 to complete this question. Download the worksheet *Human evolution map*. On the map, indicate the most likely path taken by our ancestors when they moved out of Africa. Show where they may have interbred with Neanderthals and Denisovans. Make sure you are able to support each decision that you make with evidence. Write this evidence on the map.

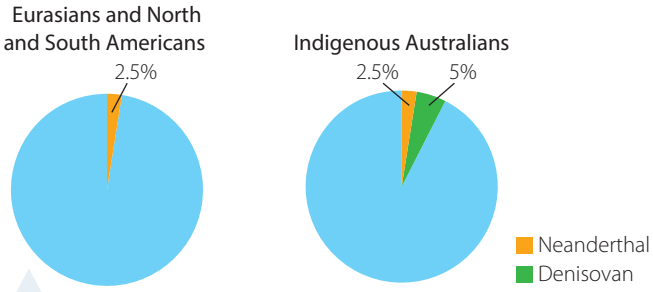


FIGURE 6.25 Percentage of archaic DNA in different human populations

Worksheet
Human evolution

KEY CONCEPTS

- Anthropological genetics is the science of using genetic data to understand human evolution.
- Humans have more genetic diversity within a population than between populations.
- Until recently, our understanding of human evolution was based on fossil evidence, which is both incomplete and subject to interpretation.
- The two models used to explain human migration are the Multiregional hypothesis (MRE) and the Replacement hypothesis.
- Genetic data favours the Replacement hypothesis that modern humans evolved out of Africa and spread across the other continents.

CHECK YOUR UNDERSTANDING 6.4

1 What does anthropological genetics aim to explain?
2 a State the two theories that attempt to explain human migration.
  b Compare these two theories.
3 Which of the two theories in Question 2 does the genetic evidence support? List that evidence and explain how it supports the theory.
4 List the two other human species that were alive at the same time as our ancestors. State the evidence that supports this.

6 CHAPTER SUMMARY

Inheritance patterns in a population: Can population genetic patterns be predicted with any accuracy?

Inheritance patterns can be studied and predicted using genetic technologies that determine the sequence of genes along a section of DNA.

Ethical issues arising from gathering of genetic information

DNA sequencing
Determines the exact order of bases of a gene, e.g. ACGTACGTACTTGGA

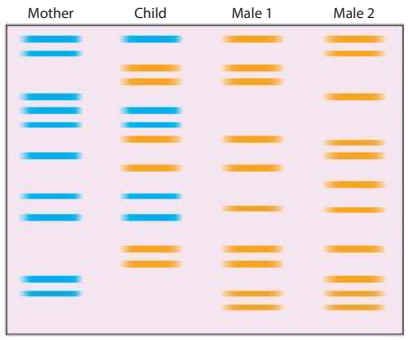
Several methods can be used, including:

Sanger method
DNA is isolated and replicated using PCR.

Maxim Gilbert method
Chemicals are used to identify a specific base. Ectrophoresis is then used to compare the patterns of bases.

New-generation technologies
(e.g. Oxford nanopore) DNA is propelled with a motor protein through a protein nanopore.

DNA profiling
Also known as DNA fingerprint analysis



STR's (short tandem repeats) are sections of non-coding DNA that are unique to individuals and can be used for the identification of individuals.

The process:
- DNA is isolated.
- PCR is used to amplify the amount of DNA.
- Sections are separated according to length using gel electrophoresis.

Gene pool
The alleles found in a population

Population genetics
The study of genetic variation in a population and changes to the frequency of alleles within a population. Data analysis from large-scale collaborative projects is used for:

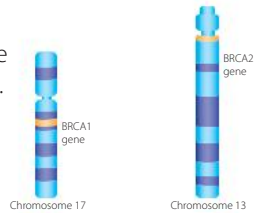
Human evolution studies
See next page.

Conservation management
How to avoid extinction by maintaining genetic biodiversity, e.g. the koala.

Traditionally field observations were used. Now genetic data is gathered to make informed decisions on biodiversity.

Disease and genetics data
Enables scientists to study patterns of genetic disease inheritance as well as focusing in improved treatment options

For example:
BRCA1 and *BRCA2* genes code for proteins that repair genes. Mutations of these genes are linked to breast cancer.



Modern genetic tools See next page.

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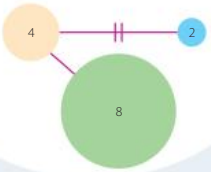
Modern genetic tools
Include SNPs GWAS and haplotypes

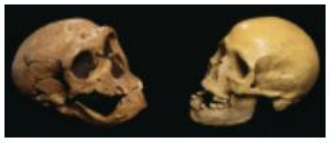
SNP
Single nucleotide polymorphism

Haplotype
A group of genes that are inherited together



GWAS
Genome-wide association study

Human evolution studies:
Anthropological genetics



Human migration theories

Modern humans of African origin conquered archaic groups and replaced them by interbreeding.

Multiregional hypothesis (MRE)
Relies mostly on fossil evidence.

Once dispersed, evolved into modern humans

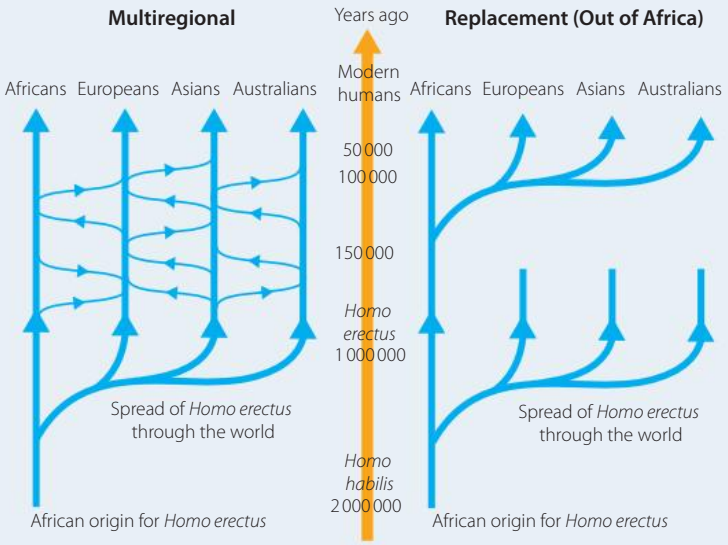
Out of Africa hypothesis
Replacement hypothesis – relies mostly on mtDNA evidence.

Suggests gene flow between neighbouring populations.

All human populations can be traced back to *Homo erectus* leaving Africa about 2 million years ago.

Archaic *Homo sapiens* left Africa.

A second migration happened about 100 000 years ago.



6 CHAPTER REVIEW QUESTIONS

Review quiz

1 a Outline two theories used to explain how humans evolved.
  b Outline the evidence used to support each theory.
2 Distinguish between Neanderthals and Denosovans. Comment on their relationship to modern humans.
3 Outline the evidence used to explain human evolution.
4 Outline how research into population genetics has changed over time.
5 How did scientists study genes and their location before the Human Genome Project (HGP)?
6 How has our understanding of human evolution changed because of large-scale data analysis?
7 Describe how DNA sequencing can assist in identifying mutations that cause diseases.
8 Explain why the HGP provided an important stimulus for the automation of DNA sequencing.
9 Outline one use of DNA profiling.
10 Describe a technology used to improve our understanding of inheritance patterns.
11 How does inbreeding affect genetic diversity?
12 a Justify why scientists have researched the genetic diversity of koalas.
  b Outline two genetic techniques used to investigate the diversity of koala populations.
  c Why might there be a difference in genetic diversity between koala populations?
13 Describe variation in the human species.
14 Explain how the frequency of an allele can change in a population.
15 Use an example to describe a genetic disease that can occur more frequently in a particular ethnic group.
16 Outline the function of the following genes:
  a tumour suppressor gene
  b BRCA1 and BRCA2.
17 How has anthropological genetics changed our understanding of human evolution?
18 Why did scientists in the past think that humans belonged to distinct racial groups?
19 Describe one phenotypic difference that is evident between human groups.
20 Explain the significance of gene flow between human populations.
21 Explain why mitochondrial DNA is used to compare the genetic relatedness of modern human populations.
22 According to the Replacement (Out of Africa) hypothesis, modern humans moved out of Africa to populate the rest of the world. Outline what may have happened to the populations that already lived there.
23 Evaluate the use of genetic techniques in aiding our understanding of human evolution.
24 Can population genetic patterns be predicted with accuracy?

Exam preparation

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MODULE 5: HEREDITY

Answer the following questions.

1 Explain why the genetic code needs to be universal in order for bacteria to produce human growth hormone. Use a flow chart to provide an example that illustrates your answer.

2 The diploid number of chromosomes in humans (*Homo sapiens*) is 46 and the diploid number of chromosomes in rice (*Ozyra sativa*) is 24. Decide whether each of the following statements is true or false, and provide evidence from your understanding of genetics to support your answer.

- **a** Simpler organisms have fewer chromosomes than complex organisms.
- **b** Plant species have fewer chromosomes than animal species.
- **c** The number of chromosomes in a human gamete is less than the diploid number of chromosomes in rice.
- **d** The number of chromosomes in a rice gamete is the same as the number of chromosomes in a human gamete.

3 The giant panda is native to China. Due to habitat loss it is on the brink of extinction. Luckily there are many giant pandas in zoos across the world where they are bred in an effort to increase the wild population. One of the problems in breeding from a small population is the reduction in genetic variation, so it is important to keep track of breeding partners and offspring.

Pandas are very difficult to breed in captivity, so most fertilisation is achieved through artificial insemination (mixing eggs and sperm in a test tube) using semen from several males. Once fertilisation occurs, it is essential that the father be identified.

One way to do this is to collect the pandas' faeces. This faeces contains DNA not only from the panda, but from the bamboo on which they feed, and from bacteria.

- **a** What techniques would be used to determine the source of DNA in the panda faeces? Describe the purpose of each technique.
- **b** What other benefit would occur from these procedures?
- **c** Zoo keepers could also use blood samples from the panda to gain DNA. Suggest one reason why they choose to use faeces instead.
- **d** Outline how scientists could keep track of breeding partners and offspring to prevent reduced genetic variation in the population.

4 Compare the physical and chemical structure of DNA and proteins, including differences in chemical composition, monomers, bonding, physical shape, primary structure and overall structure.

5 In 1940, a blood cell protein was discovered in humans that had first been discovered in rhesus monkeys. The allele of the gene that produces the protein is called Rh positive (Rh+) and is dominant over the allele for the absence of the protein, termed Rh negative (Rh^-). The allele for Rh+ shows complete dominance. Draw one Punnett square to show all of the following, and explain each using examples from the Punnett square.

- **a** An Rh^- child is born from parents who have at least one Rh^- allele.
- **b** Two Rh^+ parents could have a Rh^- child.
- **c** Siblings in the same family are a mixture of Rh^+ and Rh^-.

6 Familial hypercholesterolaemia is a disorder in which receptors on liver cells that normally take up cholesterol from the bloodstream do not work. This results in very high levels of cholesterol in the blood. Individuals who are heterozygous have half of the receptors functioning; homozygous individuals have no receptors functioning.

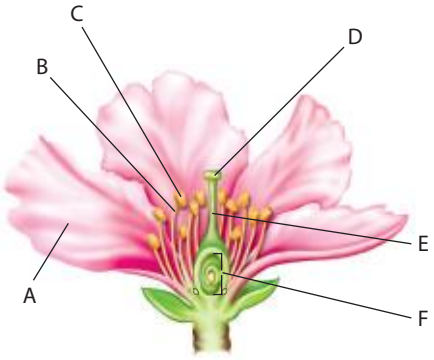
- **a** Name the type of inheritance pattern shown in hypercholesterolaemia, giving reasons for your answer.
- **b** Describe the expected effect of the gene for hypercholesterolaemia on cholesterol levels in the blood of a person who is:
  - **i** homozygous for the normal allele
  - **ii** heterozygous
  - **iii** homozygous for the hypercholesterolaemia allele.
- **c** Draw a Punnett square to determine the genotypes of offspring of two parents who are heterozygous for the hypercholesterolaemia gene. Give the phenotypic and genotypic ratios of offspring.

7 A male guinea pig that is pure breeding for black fur is crossed with a guinea pig that is pure breeding for white fur. The offspring have parts of their body that are black and parts that are white.

- **a** What type of inheritance does this suggest for coat colour in these guinea pigs? Give a reason for your answer.
- **b** Discuss what the expected offspring would be in terms of genotypes and phenotypes (and their ratios) if, in guinea pigs,
  - **i** black was dominant over white
  - **ii** black and white showed incomplete dominance
  - **iii** black was X-linked and dominant
  - **iv** black was codominant and sex-linked.

8 Discuss four natural mechanisms of reproduction in living organisms that ensure the continuity of the species. Include in your discussion examples of mechanisms seen in both asexual and sexual reproduction and unicellular and multicellular organisms.

9 Use the diagram below to answer the questions that follow.



a Give the letter(s) of the label that points to the part(s) of the flower where each of the following occurs:

i meiosis

ii pollen lands during pollination

iii fertilisation.

b Explain how artificial pollination is carried out, referring to letters of parts of the diagram to help you explain.

c In terms of the diagram, explain the difference between an ovary, an ovule and an ovum. You may redraw part of the diagram and label it to aid your explanation.

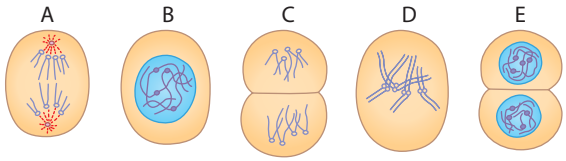
d If a harmful mutation occurred in a cell at position A, explain the consequences, if any, for this plant and future generations.

e If a mutation occurred in the cells inside C, explain the possible consequences for this plant and future generations.

f Explain how a flower such as this one could ensure cross pollination rather than self-pollination, and the resulting advantages to future offspring.

g If the inheritance pattern of this flower's colour is incomplete dominance, and it reproduces by self-fertilisation, predict the genotypic and phenotypic ratios of offspring in the first filial generation.

10 The sequence of diagrams below represents the stages of a cell dividing by mitosis.



a Write out the letters in the order in which these stages occur.

b How many chromosomes are there in the cell?

c What would the haploid number of chromosomes be for these cells?

d Draw the cells that would result if the above cell was to divide by meiosis. You may use colour to help you illustrate this process.

11 A species of mosquito that causes yellow fever has three pairs of chromosomes in its cells. Calculate how many different combinations of maternal and paternal chromosomes would be possible in the gametes. Show your working.

12 An inherited condition known as ichthyosis results in scaly skin. The disorder is found in 1 in 6000 males, but the disease is almost unknown in females.

a What type of inheritance may account for the difference in the occurrence of ichthyosis in males and females?

b From which parent would an affected male inherit the defective allele? Explain why.

c What is the probability that a male could pass the defective gene to his sons? Explain your answer.

d What may account for the fact that there are no known cases of the disease in females?

13 A dingo is a carnivorous placental mammal with high-quality parental care. The Tasmanian devil is a carnivorous marsupial mammal. It is not known exactly how many chromosomes a thylacine tiger had, but scientists expect it to be fourteen, like related marsupials.

Scientists believe that by placing synthetic chromosomes created from DNA fragments from a preserved Tasmanian tiger genome into a treated egg cell of a related species, it may be possible to create a viable egg that can be implanted into a surrogate mother, to recreate the extinct Tasmanian tiger.

Which host animal, dingo or Tasmanian devil, would be best to use to act as an egg donor? Which would be best as a surrogate mother? Justify your answer.

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DEPTH STUDY SUGGESTIONS

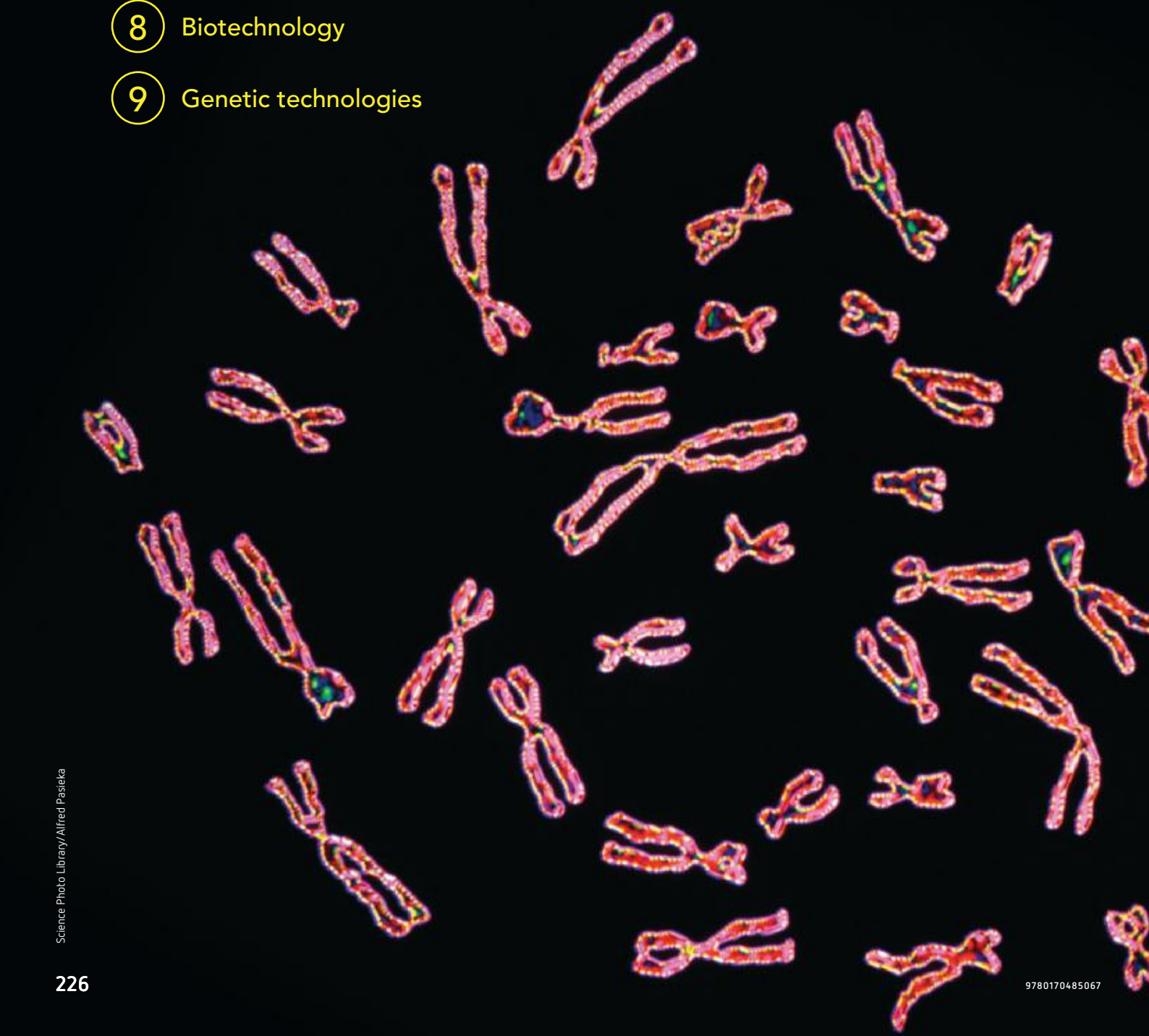
- Investigate the structure and physiology of a variety of flowering plants, with an emphasis on reproductive mechanisms that are adaptations that ensure continuity of the species.
- Research the claim that sexual reproduction helps speed up evolution and may allow some algae to adapt quickly enough to tolerate the rise in sea surface temperature. Evaluate the possibility that this adaptation will allow some corals to survive a bleaching event.
- Investigate gametogenesis and compare and model differences in meiosis during the production of egg cells and sperm cells in mammals.
- Look into the Red Queen hypothesis and find out whether it promotes natural selection for or against sexual reproduction.
- Find out about current research into links between:
  - oestrogen and cognitive functioning, sex drive, mood lowering (depression), binge eating, bone growth and maturation, maintenance of the cardiovascular system, immune system and other metabolic functions
  - androgens and muscle mass, the brain and behaviour, and the female body.
- Find out about the effects of alcohol consumption on hormonal secretions in pregnancy and the development of the foetus.
- Find out about babies conceived using DNA from three people to bypass mitochondrial disease. Use the weblink to help you.

Weblink
Three parents

- Find out about the research that has led to our understanding of how fertilisation occurs, including the research of scientists such as Oscar Hertwig, Walter Sutton and Theodore Boveri, Frank Lillie, Benjamin Kaupp, K. Miki and D.E. Clapham.
- Conduct secondary source research into the first baby born by in vitro fertilisation (Edward and Steptoe technique) in 1978 and compare this technique to the techniques used today.

» MODULE SIX

GENETIC CHANGE



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7 Mutation

INQUIRY QUESTION

How does mutation introduce new alleles into a population?

Students:

- explain how a range of mutagens operate, including but not limited to: ICT
  - electromagnetic radiation sources
  - chemicals
  - naturally occurring mutagens
- compare the causes, processes and effects of different types of mutation, including but not limited to: ICT N
  - point mutation
  - chromosomal mutation
- distinguish between somatic mutations and germ-line mutations and their effect on an organism (ACSBL082, ACSBL083) ICT
- assess the significance of 'coding' and 'non-coding' DNA segments in the process of mutation (ACSBL078) ICT N
- investigate the causes of genetic variation relating to the processes of fertilisation, meiosis and mutation (ACSBL078) N
- evaluate the effect of mutation, gene flow and genetic drift on the gene pool of populations (ACSBL091, ACSBL092) N

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
- **7.1** Secondary-source investigation into mutagens and how they operate
- **7.2** Analysing data to investigate mutations in coding and non-coding DNA
- **7.3** A secondary-source investigation of the causes of mutation related to meiosis and fertilisation

Worksheets
- Mutagens and their effects
- How the term 'mutation' was introduced into Biology
- The biochemical nature of mutations
- Introducing the Hardy-Weinberg equation

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

FIGURE 7.1 Mutations at the molecular level alter the sequence of nucleotides in DNA.

Our genes play a big part in determining how we look and behave, and even whether or not we are likely to get sick. No two individuals are alike – even identical twins have their differences. These differences are termed 'variations'. The combined impact of genetics and the environment is being explored to find out what makes us different. Mutations, or changes in DNA, play a large role in introducing new alleles into the population. Scientists are using knowledge about variations due to mutation in exciting new ways, such as predicting and treating health problems.

At the molecular level, a **mutation** is a change in the genetic material of a cell – the sequence of nucleotides in DNA is altered. This may be the result of spontaneous mistakes that arise and are not corrected when DNA is replicated, or of mistakes induced by environmental factors such as UV light or cigarette smoke. In either case, this alteration of DNA changes the information coded in the gene and may alter the protein or RNA end product.

Mutations range in size, from a change in a single DNA base pair to a change in the structure of a large segment of DNA in one or more chromosomes, where multiple genes are affected. Variations of genes that arise as a result of mutations are said to be alleles of that gene. Some genes have markers that may be associated with mutations, and data about these markers is being used more frequently to identify potential health problems.

7.1 Mutagens

What is a mutagen and what are its effects? Answers to these questions were discovered just over a hundred years ago. During the late 1800s and early 1900s, many scientists were involved in studying radiation. Because the harmful effects of radiation were unknown then, scientists who were exposed to large amounts of radiation over prolonged periods of time, such as Marie Curie, developed various illnesses. Marie Curie worked with ionising radiation for most of her career and died in 1934 from leukaemia due to overexposure to radioactive emissions. Rosalind Franklin, who worked with X-rays in her crystallography studies, died of ovarian cancer in 1958.

Survivors of the 1945 bombing of Hiroshima suffered physical mutations as a result of radioactive output from the nuclear explosion. The link between exposure to ionising radiation and an increase in the occurrence of certain illnesses such as leukaemia and other cancers was identified, but further evidence was needed to show that radiation was causing these cancers.

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By the 1970s, it was known that certain chemicals as well as radiation can change the structure of DNA and lead to cancer. Victims of the 1986 nuclear meltdown in Chernobyl showed immediate damage to their DNA and the damage was passed to their descendants. As early as the 1950s, some viruses were found to be carcinogenic. So how do chemicals, radiation and biological agents change DNA in a way that leads to cancer?

Mutagenic agents

Maintaining the integrity of DNA is essential for the functioning of cells. Environmental agents that alter DNA and cause mutations are termed **mutagens**. The process of inducing a mutation is termed **mutagenesis** and the resulting mutations are termed **induced mutations**.

Many mutagens are **carcinogenic** (cancer-causing). This is because some mutations occur in genes that regulate the cell cycle or promote or suppress cell division. These mutations cause changes in the cell cycle that may result in increased cell division with no differentiation, resulting in masses of cells known as tumours. There are two types of genes in which mutations commonly lead to cancer: proto-oncogenes and tumour suppressor genes. You will learn more about these in Chapter 15.

Mutagens can be grouped into categories, based on their source (Fig. 7.2). To explore different mutagenic agents, you need to understand their basic features, how they act and the specific damage to DNA that commonly results from each type of mutagen.

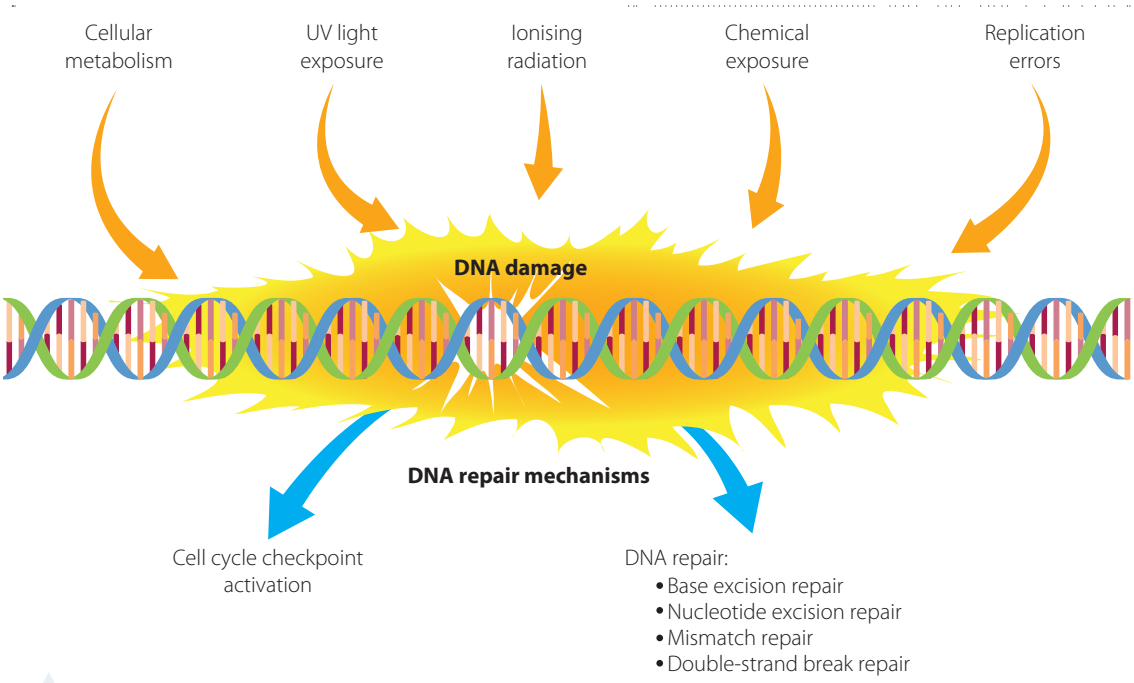

FIGURE 7.2 Causes of DNA damage and types of repair mechanisms activated

Chemical mutagens

Chemical mutagens are chemicals that cause mutations if cells are exposed to them at high frequencies or for prolonged periods of time. A large number of ingested and everyday chemicals have been found to be mutagenic over time, and many of these are no longer as widely used as they were in the past. Chemical mutagens cause a change in DNA that alters the function of proteins and, as a result, cellular processes are impaired.

Examples of chemical mutagens:

- *ingested chemicals* including alcohol, tar in tobacco smoke, some medications and chemicals in food – especially charred and fatty foods, and food additives and preservatives (such as nitrites)
- *environmental irritants and poisons* such as organic solvents (for example, benzene), cleaning products, asbestos, coal tars, pesticides and some hair dyes.

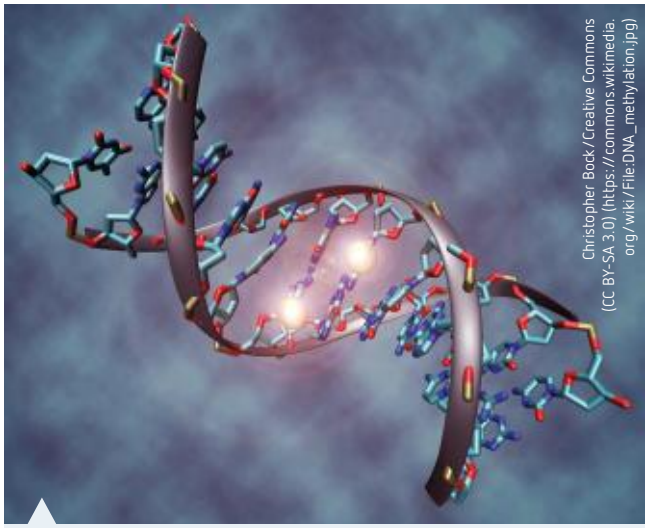

FIGURE 7.3 DNA undergoes structural change in base pairing as a result of mutagenesis.

Chemicals that are mutagenic are usually structurally similar to normal bases in DNA (purines and pyrimidines) and so they may mistakenly become incorporated into DNA during replication. This results in the insertion of incorrect nucleotides opposite them during replication – termed **mispairing** (Fig. 7.3). Their insertion often results in the production of a non-functional protein end product. Some common examples of molecules that are chemical mutagens include alkylating agents, de-aminating agents and intercalating agents (chemicals that interact directly with DNA and change its structure). You will research this in more detail in Investigation 7.1.

Naturally occurring mutagens

It is believed that some mutations, thought in the past to have arisen spontaneously, may be the result of exposure to naturally occurring mutagens in the environment. We know that spontaneous mutations, such as DNA replication errors, arise during DNA replication and are retained because the normal mechanism of DNA repair does not correct them. Other types of mutations, perhaps thought to be 'spontaneous' in the past, are now associated with mutagenic agents that are present naturally in the environment.

Naturally occurring mutagens are mutagenic agents that are present at normal levels within natural environments, and may cause mutations. The likelihood of mutation is thought to increase with increased frequency and length of exposure.

Naturally occurring mutagens can be divided into two groups: **biological mutagens** and **non-biological** naturally occurring mutagens.

Non-biological naturally occurring mutagens include metals, such as mercury and cadmium, that occur naturally in the environment. *Biological* naturally occurring mutagens include viruses, bacteria, fungi and their products.

Biological mutagens and their actions

- *End-products of metabolism:* many naturally occurring biological mutagens may be produced by fungi, or plant or animal cells during metabolism. These mutagens tend to be discovered when sudden outbreaks of particular types of cancers occur in organisms that live in particular areas or soils and/or eat particular foods.

  One example of a biological mutagen that is currently being researched is an end-product of cellular metabolism, called *nitrosamine*. Nitrosamines are chemicals that form in the stomach when certain foods or food ingredients are eaten in combination – for example, when ingredients that contain nitrous acid or nitrites are eaten together with amines, naturally present in meat and fish. Examples are some processed and smoked meats and sausages that have this combination of chemicals. Cooking these foods at high temperatures causes the nitrites and amines to combine, forming carcinogenic (cancer forming) nitrosamines, particularly if the cooking methods involve

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direct heat, such as frying or grilling. (It is interesting to note that nitrosamines are a component of cigarette smoke, an unnatural mutagen that is known to be highly carcinogenic.)

- **Transposons** are sections of DNA that spontaneously fragment and relocate or multiply within the genome. When these transposable elements insert into chromosomal DNA, they disrupt DNA functioning.
- *Microbes* are naturally occurring biological mutagens. These include *viruses* (such as hepatitis B virus, HIV, Epstein-Barr virus, Rubella virus) and *bacteria* (such as *Helicobacter*). Mutagenic microbes may also directly alter the genetic material in cells.

Effects of biological mutagens

Many mutagenic microbes are able to insert their own base sequences into DNA and in this way change the functioning of genes and trigger cancers (similar to transposons).

Some bacteria and/or their products cause inflammation, during which free radicals (reactive oxygen species) are produced, causing DNA damage and reducing the efficiency of DNA repair systems, thereby increasing mutation. Examples are the bacteria *Helicobacter pylori* and some mycotoxins (chemicals produced by some fungi). The release of free radicals in the body may also lead to a condition known as oxidative stress, where the individual's immune response is altered and the immune system does not function properly to fight off viral infections.

Some products made by microbes are mutagenic due to their instability at cellular pH. They decompose to form an intermediate that can bind to cellular DNA and alter it.

Physical mutagens

Physical mutagens include *heat* and *ionising radiation*. Direct heat often has a combined action with chemical and naturally occurring mutagens, as discussed in the previous section.

Radiation is any transfer of energy through space from a source, such as electromagnetic radiation from the sun. Not all radiation is harmful to our health. The harmful type, called ionising radiation, has enough energy to break chemical bonds in molecules, including DNA.

Electromagnetic radiation

Electromagnetic radiation comes from the sun and is a form of energy that is all around us, such as radio waves, microwaves and gamma rays. This energy is transmitted in waves or particles in a range of wavelengths and frequencies.

The overall range of wavelengths is known as the **electromagnetic (EM) spectrum**. There are seven regions in the spectrum, in order of decreasing wavelength (and increasing energy and frequency): radio waves, microwaves, infrared (IR), visible light, ultraviolet (UV), X-rays and gamma-rays (Fig. 7.4).

Ionising radiation includes the shorter wavelengths of UV radiation as well as X-rays and gamma rays. The shorter wavelength and high energy of ionising radiation make it dangerous, as it can split off electrons, which cause damage in cells. Radio waves and infrared radiation are long-wavelength, low-energy forms of radiation and are not harmful; ultraviolet radiation is somewhat harmful; gamma radiation is extremely harmful, even in small doses.

Ultraviolet (UV) radiation

Near-ultraviolet radiation or UVA (315–400 nm) is non-ionising and the DNA damage that it does may be related to ageing, but its mutagenic and carcinogenic impact is still uncertain.

Artificial UV lights in tanning salons use UVA, and prolonged exposure is a risk to health. For this reason, commercial tanning salons are banned in Australia.

UVB or middle UV (280–315 nm) and UVC or far UV (180–280 nm), which have shorter wavelengths, are a form of ionising radiation that is high in energy and the chemical damage it causes to DNA by breaking bonds is known to be mutagenic and carcinogenic.

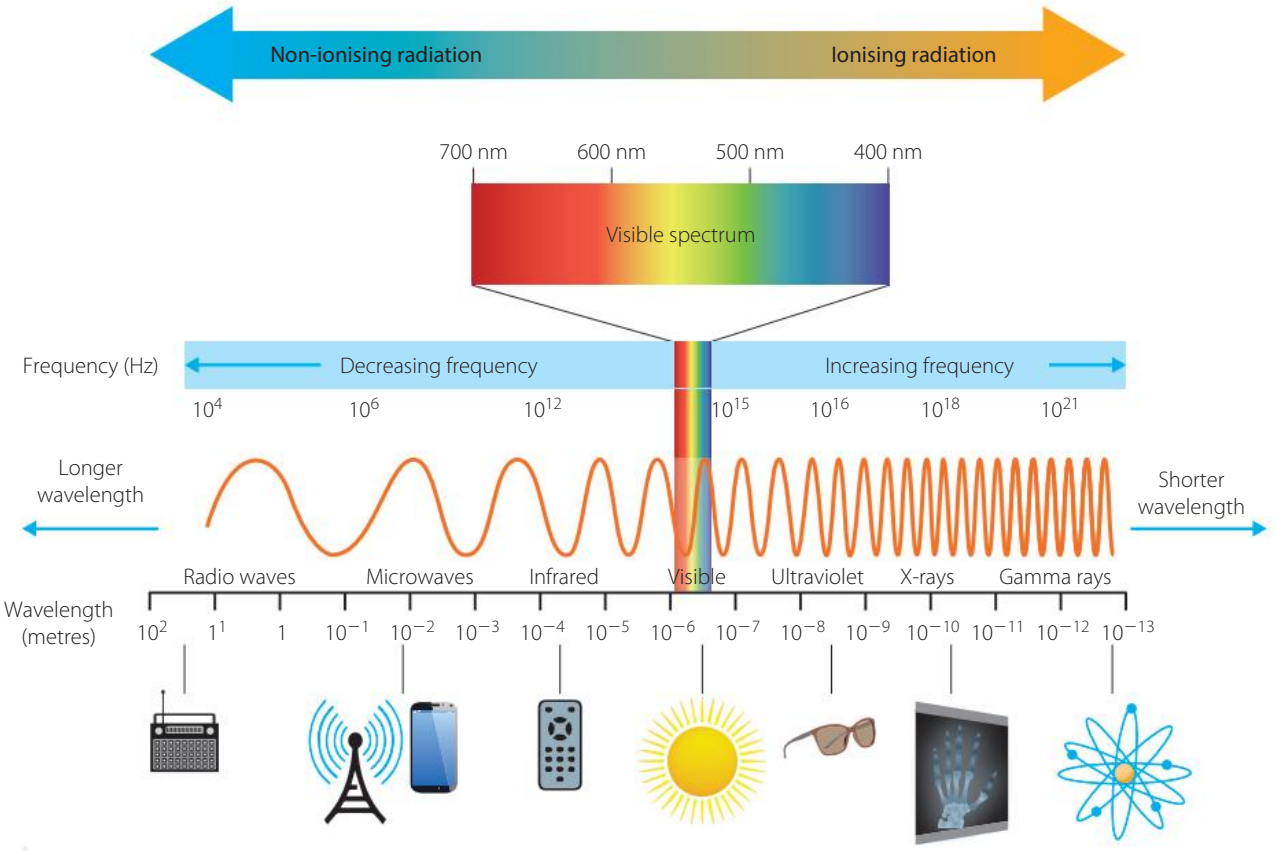

FIGURE 7.4 Ionising and non-ionising radiation

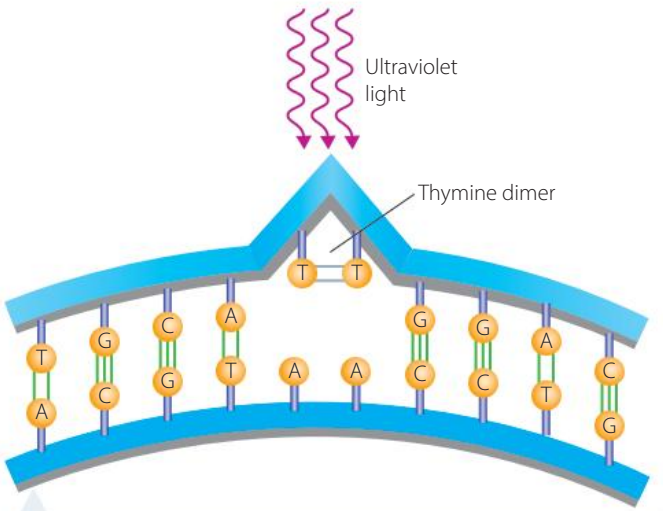

FIGURE 7.5 Thymine dimer mutation caused by exposure to UV light

Naturally occurring UV radiation from sunlight has been shown to be a contributing factor to skin cancer. The most common effect of UV radiation on DNA is to produce pyrimidine *dimers* (cross-linked nucleotides). This occurs when an adjacent pair of bases (either two thymine or two cytosine bases) on the same strand become attached to each other (Fig. 7.5). This prevents them from pairing with bases on the complementary strand, causing the strand to end prematurely. This prevents normal replication and transcription, affecting both the cell cycle and gene products.

Other forms of ionising radiation

Ionising radiation is high-energy radiation that is able to free electrons from atoms or molecules, turning them into ions (charged particles). These high-energy electrons that are released can damage DNA.

Sources of ionising radiation are cosmic rays from the sun and outer space, as well as radioactive elements in soil, the atmosphere and even stone and wood. Artificial sources of radiation that have been created by humans include radioactive materials from nuclear reactions. These emit radiation such as alpha, beta and gamma rays. Radiation from atomic bombs, toxic spills (such as occurred at Chernobyl in 1986), nuclear testing and power plants, and even radiation used in medicine (such as X-rays and gamma radiation) are all forms of mutagenic ionising radiation that can be harmful to human health, even in

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small doses. As mentioned before, high-energy UV radiation may also be considered ionising radiation, if it has enough energy to ionise (remove an electron from) an atom or a molecule.

Ionising radiation has many effects on DNA. When electrons pass through cells, they interact with water, and particles called free radicals are released (Fig. 7.6). These are highly reactive and may act on proteins and lipids in cell membranes and other parts of cells, in addition to DNA. Breaks in one or both strands of DNA occur, leading to deletions, partial chromosome loss, rearrangements of sequences in DNA and cross-linking of DNA with itself. This interferes with cell division, gene products and cell metabolism.

Weblink
How radiation changes your DNA
Watch the video clip.

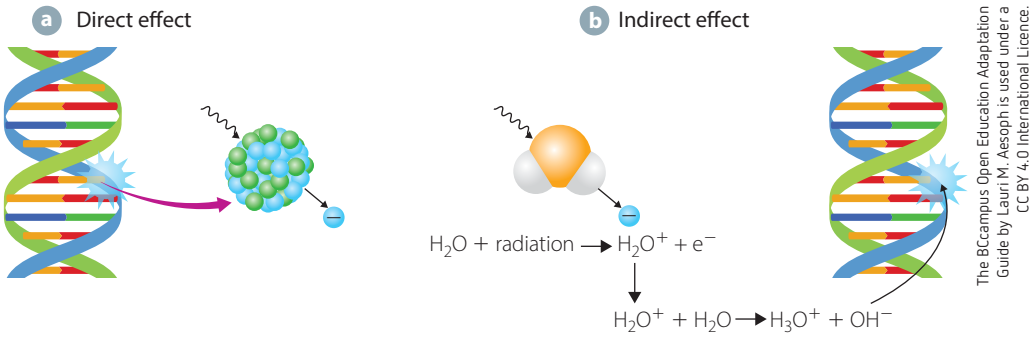

FIGURE 7.6 The effects of ionising radiation on DNA: **a** release of an electron from a DNA molecule; and **b** formation of free radicals, which can affect DNA structure

See Chapter 3, page 104, for further details on DNA repair.

Today there is strict regulation of the quantity of mutagens that may be present in products or to which people may be exposed. Some chemicals are banned and others may be added in only very small amounts. Radiation doses are also regulated.

Because the integrity of DNA replication is so important, DNA repair mechanisms operate in cells, whereby enzymes that are involved in replication also play a role in removing damaged parts of DNA and repairing DNA. These mechanisms include:

- base excision repair – a damaged or incorrectly paired base is removed (by a nuclease enzyme) from its sugar linkage and replaced; an example is the removal of a pyrimidine dimer (Fig. 7.7).
- mismatch repair – once DNA has replicated, the enzyme DNA polymerase carries out a 'spell check' for accuracy of replication.

Weblink
DNA damage
Watch the video clip to see how DNA exposed to ionising radiation is damaged and repaired.

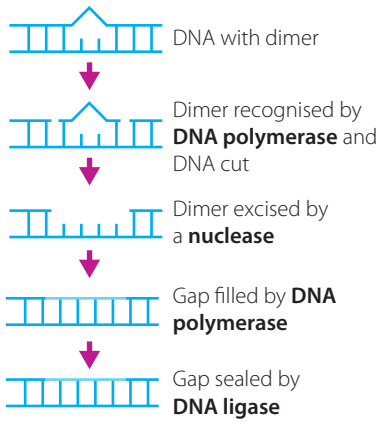

FIGURE 7.7 Repair of a pyrimidine dimer in DNA

INVESTIGATION 7.1

Secondary-source investigation into mutagens and how they operate

Worksheet
Mutagens and their effects

How do different agents alter the structure of DNA and affect gene expression? What increases the harmful effects of mutagens on DNA? Is it the intensity or dosage of the agent, the length of time the cells are exposed to the agent, or the frequency at which cells are exposed? It is important to find out such information, so that lifestyle changes can be made to reduce the rate of mutagenesis.
There are two parts to this investigation. Part A is undertaken in a group of no more than four members and is mandatory. Part B is an optional extension.

Information and communication technology capability

Literacy

Critical and creative thinking

AIM

To investigate mutagens and how they operate

PART A: TYPES OF MUTATIONS CAUSED BY SPECIFIC MUTAGENS

METHOD

1. Investigate the specific chemical changes that are induced in DNA by each type of mutagen: radiation, chemicals and naturally occurring mutagens such as biological agents. Use specific examples to support your answer.
2. Working in groups, each member might choose to investigate one type of mutagen and its effects on DNA and then share results.
3. Decide as a group exactly what information you will need to gather (develop a set of research questions) and how you will present your results. You should include illustrations or images (correctly referenced) of the type of DNA damage caused by each mutagen. Refer to tables in the online resource to assist you with your research. You may also wish to include information on safe exposure levels and/or levels at which the agent becomes mutagenic.
4. Some suggestions for presentation of results are: in the form of a table, as a multimedia presentation, or in a poster. Select the presentation style that will best enable you and your group to understand and revise mutations.
5. Find a video clip of not more than 5 minutes that shows how mutations arise. Ensure you use a reliable source. Save the URL and record it with your results. Write a paragraph to justify the reliability of your source.
6. Pair and share with a partner and view each other's selected video clips. Review them and rate them on a scale of 1 to 5. You and your partner should develop criteria on which you will rate the videos.

Review how to develop research questions and assessing the reliability of resources in Chapter 1

RESULTS

1. Present your information in the format chosen by your group. Make sure all research questions are addressed, examples are used and an illustration of the type of DNA damage is included.
2. Create a document in which you record:
   - the URL for your video clip
   - the title of the video
   - a brief synopsis of the video (See margin note.)
   - a paragraph justifying the reliability of the source
   - a place for your partner to write a paragraph as a review of your video clip.

A synopsis is a short description of what happens in a film. It includes the most important or interesting parts, to attract a potential target audience.

DISCUSSION

Do a peer review: evaluate the relevance and reliability of your partner's video clip.

CONCLUSION

Write a conclusion about mutagens and how they operate.

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PART B (OPTIONAL EXTENSION): WHAT INCREASES THE RATE OF MUTAGENESIS?

Numeracy

METHOD

1. Using a range of reliable sources, research whether it is the length of time that cells are exposed to mutagens, the frequency of exposure (how often they are exposed), the intensity (dosage) of the exposure to the mutagen, or any combination of these factors, that leads to an increase in the harmful effects of mutagens on DNA.
2. Write a literature review with an investigation question and three to four paragraphs summarising your findings. Include graphs to support your viewpoint.
3. Propose a hypothesis and design a practical investigation, based on your secondary-source research, that you could carry out to test your hypothesis.

   Note: For safety reasons, it is not recommended that a practical investigation of this kind be conducted, as the use of mutagens poses risks. If you are going to conduct a practical investigation, use common household products that do *not* have highly mutagenic effects, and do so only with the approval of your teacher once you have considered all safety aspects with them.

Review how to write a literature review in Chapter 1

RESULTS

1. Your literature review, with graphs, should summarise what factors increase the rate of mutagenesis.
2. Your experiment design should be written as a procedure with the headings Aim, Hypothesis, Materials, Risk assessment and safety, Method and Results (even though you will not have any results to record at this stage). In the section on results, draw up a scaffold for recording your results – for example, a table with headings and/or a graph with axes labelled.

DISCUSSION

Do a peer review of a classmate's experiment design, and provide feedback on how they could improve their investigation plan.

KEY CONCEPTS

- A mutation is a change in the nucleotide sequence in DNA.
- Mutations may arise spontaneously during cell division or they may be induced by mutagens.
- Physical mutagens include various types of radiation that cause DNA damage.
  - UV light causes dimers (cross-linked nucleotides).
  - X-rays cause chromosomal aberrations.
  - Nuclear radiation causes breaks in DNA strands.
- Chemical mutagens include ingested chemicals (such as tar in tobacco smoke) and irritants or poisons (such as benzene). Chemical mutagens may disrupt DNA by replacing bases or being inserted between them.
- Naturally occurring mutagens include biological and non-biological naturally occurring elements in the environment.
- Biological mutagens include some bacteria that insert plasmids into DNA and some viruses that insert their nucleic acid sequences into DNA.

CHECK YOUR UNDERSTANDING 7.1

1. Distinguish between 'mutation' and 'mutagen'.
2. Name the three types of mutagenic agents and give two examples of each.
3. Define 'naturally occurring mutagen' and give an example.
4. Explain how DNA replication is disrupted by:
   - **a** dimer formation
   - **b** free radical interference
   - **c** single-stranded breaks
   - **d** double-stranded breaks.
5. Discuss the importance of DNA repair enzymes during replication.

7.2 Types of mutations

Genes owe their specificity to the order in which their base pairs are arranged (in much the same way as words derive their meaning from a specific sequence of letters in the alphabet).

Mutation is a collective term for a change in DNA, but the different types of mutation can be distinguished according to five criteria:

See Chapter 4 for discussion of introns and exons.

1 *The origin (cause) of the mutation:* **spontaneous mutations** arise randomly as a result of an error in a *natural process* such as DNA replication in cells, whereas **induced mutations** arise as a result of an *environmental agent* such as a chemical or radiation, which increases the chance of nucleotide sequences being changed.

2 *The amount of genetic material changed:* **point mutations** are changes to a single base pair of DNA and affect only a single gene (**gene mutations**). In contrast, chromosomal mutations move whole blocks of genes to different parts of a chromosome or to another chromosome entirely (Fig. 7.8). **Frameshift mutations** may affect a single gene or a sequence of genes and arise as a result of a point mutation or a chromosomal mutation (Fig. 7.11, page 239).

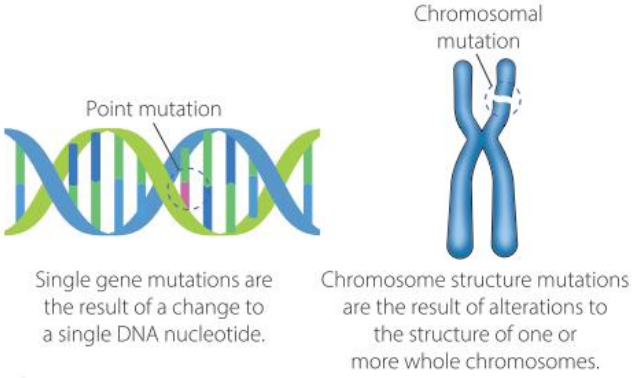

FIGURE 7.8 Mutations range in size from single gene mutations to chromosomal mutations.

3 *The effect of the mutation on DNA:* a nucleotide base may be substituted, inserted or deleted. This in turn may lead to a change in one amino acid (or no change in amino acids if the new base forms a triplet that codes for the same amino acid as the original codon).

4 *The effect of the mutation on phenotype:* there may be no change in the phenotype (silent mutation), or a small or large change (or variation) in the phenotype, depending on the type of amino acid substituted. If a phenotypic change occurs, this is most often *harmful*, but it may sometimes be *neutral* or, very rarely, *beneficial* in its effect on the individual and its chances of survival.

Mutations that occur in genes that code for proteins are usually extremely severe, with many being potentially lethal for the organism that bears them.

5 *Heritability of mutations:* the possibility that a mutation will be passed down through generations depends on whether the mutation occurs in a non-reproductive (somatic) cell or a reproductive (germline) cell. Mutations that occur in non-coding DNA may or may not have severe effects, depending on whether the DNA sequence is involved in gene regulation.

Point mutation

A point mutation is a single nucleotide variation. Although small, these mutations can have a significant effect on phenotype if they occur within the exon of a gene, or in an intron where they affect gene expression.

Weblink
GIF animation of a substitute mutation
Watch the animation, take notes and draw diagrams to consolidate your understanding.

Effects of point mutations on DNA

Point mutations may be named according to how they change the nucleotide sequence of DNA. This in turn affects the expression of the mutated genes. Most point mutations result in a base substitution, but some result in a frameshift mutation. (See next section.)

A base substitution occurs when one nucleotide base is replaced by a different base – for example, C is replaced by T, G or A (Fig. 7.9). This may result in a different amino acid being inserted into the polypeptide.

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As an analogy, think of the bases on DNA as being like letters in words. If the word TAG becomes GAG, changing this word in a sentence will give the sentence a different meaning. In the same way, changing an amino acid in a protein can change the function of the protein.

An example of a point mutation is the sickle cell gene point mutation that occurs in human red blood cells, resulting in the disease sickle cell anaemia. The triplet CTC is changed to CAC (and the complementary strand changes from GAG to GTG). In terms of its phenotypic effect, it is termed a 'missense mutation'. The base substitution causes the amino acid glutamate to be swapped for valine, alters the shape of the haemoglobin molecule and results in the sickle cell shape of the red blood cells. (See Fig. 7.10b for a visual representation of a missense mutation.)

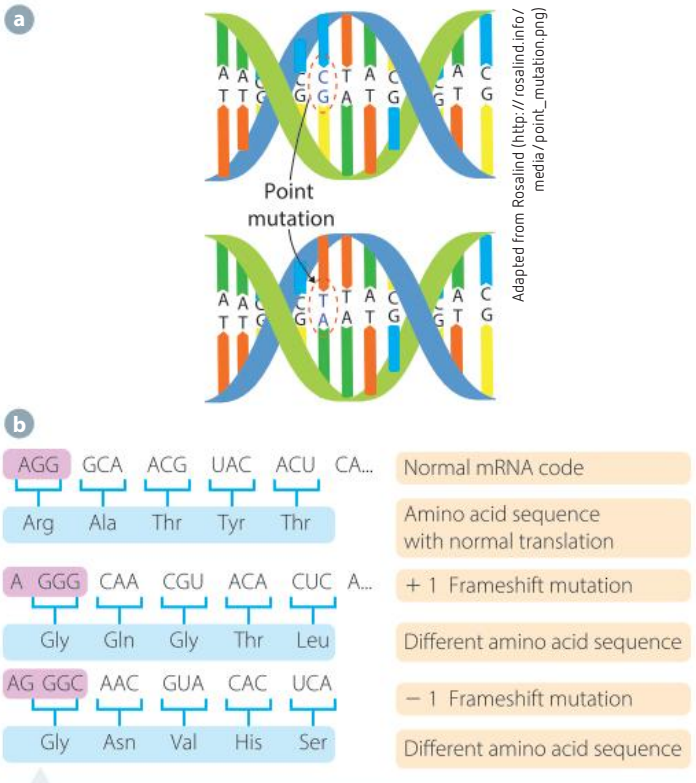

FIGURE 7.9 **a** Point mutation due to a base substitution in DNA, showing replacement of CG pair with TA; **b** frameshift mutation (shift in reading frame highlighted in pink)

Frameshift mutation

A point mutation that involves the insertion or deletion of a single nucleotide pair can lead to a frameshift mutation (Fig. 7.9). This is where the insertion or deletion of one base shifts the entire 'reading frame' of RNA, leading to the creation of a whole sequence of incorrect amino acids and the production of a non-functional protein. Because mRNA bases are read in threes (triplets of bases called codons), when a base pair is added or removed from DNA, it shifts the reading frame and every triplet beyond that point will be different (Fig. 7.10d and e).

As an analogy, think of triplets in mRNA as being like three-letter words in a sentence. A base insertion into 'THE DOG SAW THE CAT' leads to 'TTH EDO GSA WTH ECA T'.

Frameshift mutations may arise not only from point mutations, but also when more than one base is inserted – see insertions and deletions, in the next section – particularly if the bases inserted are not in multiples of three, as this will shift the reading frame (Fig. 7.10d and e).

Changes in proteins (phenotype) due to point mutations

Mutations may also be classified according to their effect on proteins and/or phenotype.

- **Nonsense mutations** (Fig. 7.10a) change an amino acid to a stop codon. This has a very noticeable effect on a protein, as it cuts it short. The resulting protein is usually non-functional and this has a major phenotypic effect.
- **Missense mutations** (Fig. 7.10b) are point mutations that result in an amino acid change. The functionality of the resulting protein is determined by whether or not the replacement amino acid is the same type as the original. The sickle cell anaemia example given earlier is a missense mutation because the protein end product is changed and is less functional (the 'sense' or 'meaning' of the protein is changed).
- **Silent mutations** (Fig. 7.10c) are changes in the DNA sequence that do not cause a change in amino acid, because of the redundancy of the genetic code, and therefore have no effect on proteins. For example, GCC and GCA on mRNA both code for the amino acid valine, and so a change between GCC and GCA in the third base of mRNA will have no effect on the amino acid sequence of a protein. Silent mutations therefore do not have any noticeable effect on the protein (or phenotype).
- **Neutral mutations** are changes in DNA that result in an amino acid of the same type as the original, and so the change does not significantly affect the structure of the protein.

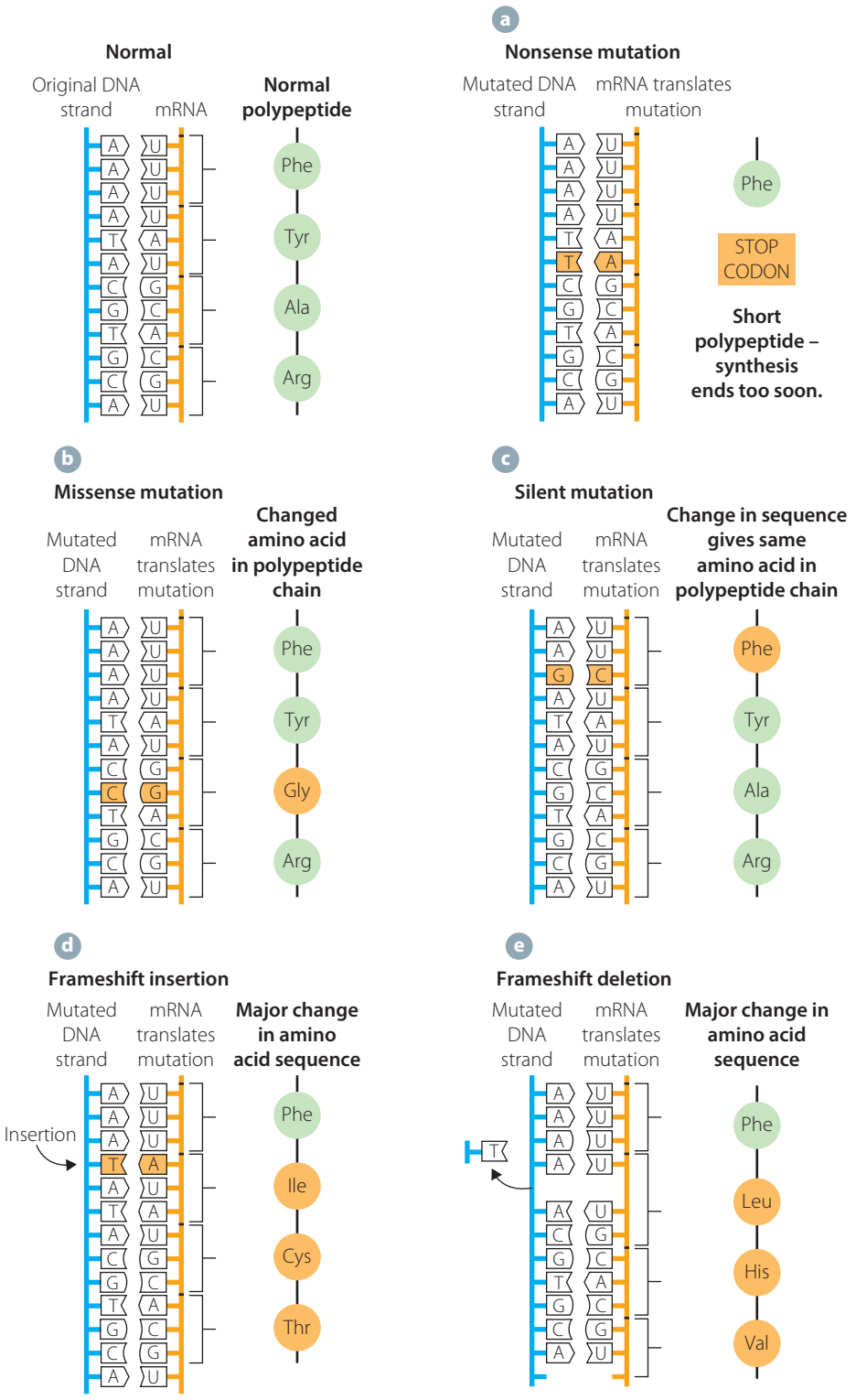

FIGURE 7.10 Types of point mutations

Chromosomal mutations

Some mutations are larger than point mutations and involve changes to a series of bases within a chromosome. Chromosomal mutations (sometimes referred to as chromosomal aberrations) are large-scale changes where either the overall *structure* of a chromosome is changed or the entire *number* of

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chromosomes in a cell is altered. Gene mutations, by contrast, are any changes to the DNA sequence within one gene, ranging from point mutations to chromosomal mutations that involve insertions and deletions of a number of bases.

Changes in chromosome structure

There are four main types of chromosomal mutation that alter the structure of chromosomes: deletions, insertions, inversions and translocations. These are outlined below and shown in Figure 7.11.

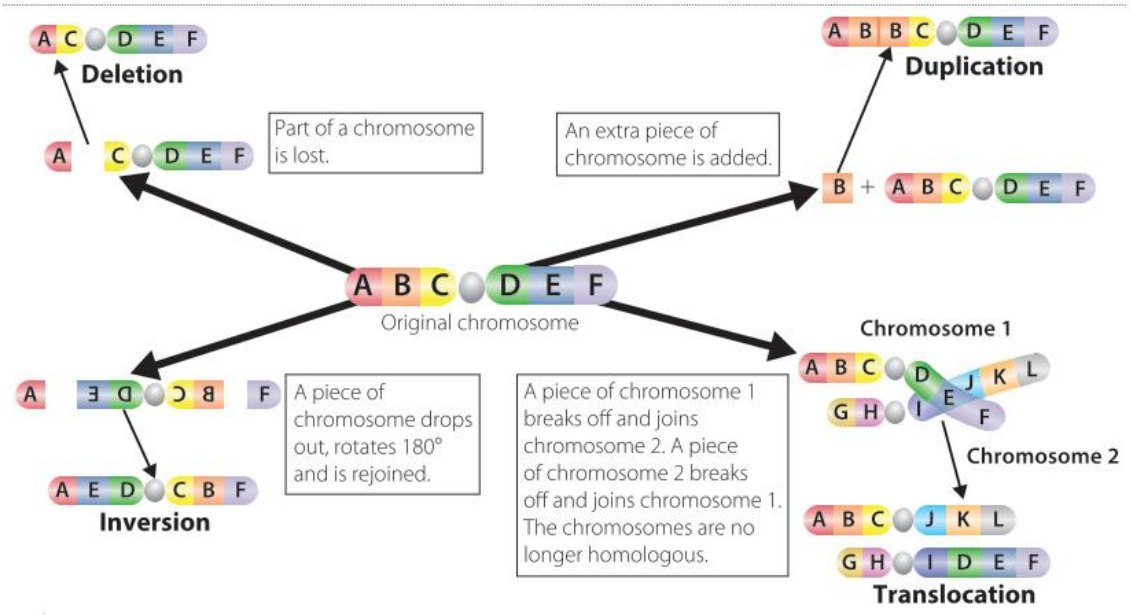

FIGURE 7.11 Types of chromosomal mutation (changes to *structure* of chromosomes)

Chromosomal deletion

A chromosomal deletion occurs when a section of DNA is removed and not replaced. This leads to a reduction in the number of genes in a chromosome. Deletions are often the result of exposure to high heat, viruses or radiation.

Chromosomal insertion (duplication)

Chromosomal insertion (duplication) occurs when a portion of DNA is duplicated (or doubled) and inserted, increasing the number of genes on the chromosome. Its effect depends on the size, location and number of repeats.

The *position* of the duplication in DNA (whether in an intron or an exon), as well as the number of repeats of the sequence, determine whether the mutation has any phenotypic effect and the extent of this effect. The *size* of the insertion or duplication also plays a role, determining whether the insertion affects the individual in the same way as altering the chromosome number does. (Changes in chromosome number are discussed later.)

Changes in the *number of repeats* may also determine the phenotypic effect. When a section of DNA is copied and repeated two or more times, the extra copies of the DNA sequence may or may not have a phenotypic effect, depending on whether the number of repeat copies is above a certain threshold number. These types of duplications lead to variations known as copy number variations. Diseases such as Huntington's chorea and fragile X syndrome seem to be linked to the number of duplications within a chromosome.

Chromosomal inversion

A chromosomal inversion occurs when a section of DNA is removed, turned around through 180 degrees (the sequence is turned back to front or inverted) and then reinserted into the chromosome, so that the bases are in reverse order. Inversions may range in size from a few hundred bases to five megabases in size (1 megabase is 1 million base pairs long). One of the most commonly known diseases that arises from an inversion mutation is haemophilia A, caused by an inversion mutation in the factor VII gene on the X chromosome. This mutation was an example used in Chapter 5 in the discussion of X-linked inheritance.

Chromosomal translocation

A chromosomal translocation occurs when a section of DNA is moved from one chromosome to a non-homologous chromosome. This rearrangement may lead to gene fusion, when the translocated region joins two normally separate genes. Some scientists think that transposable elements (transposons or 'jumping genes') inserted into DNA millions of years ago may be responsible for making up a large proportion of non-coding DNA.

Aneuploidy: changes in chromosome number

Aneuploidy occurs when one or more extra copies of an entire chromosome are made or an entire chromosome is missing, leading to an abnormal number of chromosomes in the cell. For example, the result of aneuploidy in a human cell may be 45 or 47 chromosomes instead of the normal number, 46 chromosomes. Down syndrome is an example of a disorder that arises as a result of a change in ploidy (chromosome number). The individual has an extra copy of chromosome 21. (Down syndrome is discussed in more detail in Chapter 15.)

The gaining of one or more complete sets of chromosomes is termed 'euploidy'. This differs from aneuploidy.

KEY CONCEPTS

- Mutations may be classified according to:
  - the amount of DNA affected (gene mutations or chromosomal mutations)
  - how they arise (spontaneous or induced mutations)
  - how they change the DNA structure (point mutations, frameshift mutations, deletions, insertions, translocations, duplications)
  - how they affect DNA functioning and resulting proteins (nonsense, missense, neutral, silent)
  - whether or not they can be inherited (somatic or germline mutations).
- A *point mutation* is a change in a single nucleotide base pair in DNA and is an example of a small-scale *gene mutation*.
- A *chromosomal mutation* is a large-scale alteration, being a structural and/or numerical change in the entire DNA strand. Some chromosomal mutations involve a change in chromosome number (aneuploidy).

CHECK YOUR UNDERSTANDING 7.2

1. Refer to Figures 7.10 and 7.11 (on pages 238 and 239) to answer the following questions.
   - **a** Describe, in your own words, the changes in DNA that occur in each type of mutation.
   - **b** Using the relevant letters of the alphabet shown in the diagrams, give a reason why each mutation has its particular name.
   - **c** Draw a diagram similar to those in Figure 7.11 to represent a chromosomal insertion of six base pairs.
2. Draw a sequence of diagrams, using colour and showing the base pairing in part of a double-stranded DNA molecule, to represent the following types of point mutations within a gene:
   - **a** base substitution
   - **b** frameshift mutation due to an insertion
   - **c** deletion mutation that does not lead to a frameshift mutation.
3. Outline what the effect on the protein will be for each mutation in parts a–c of Question 2.

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7.3 Mutations and how they affect organisms

A mutation is often defined in molecular terms as a change in DNA nucleotide sequence, but mutations exert their effects at cellular, individual and population levels.

At a *cellular level*, the type of cell affected by a mutation determines the extent of its influence. A mutation that affects a germline cell will be passed on to every cell in the offspring of the gamete, whereas a somatic mutation (mutation in a non-reproductive body cell) may lead to a localised effect, such as the development of a tumour in a part of the organism, but it will not be passed on to the next generation.

Mutations within *individuals* differ in their phenotypic effect. To understand mutations at the level of the individual, we need to understand how genes translate into physical, behavioural, physiological or biochemical changes in features (Chapter 4). Some mutations lead to significant phenotypic change, whereas others have little or no phenotypic effect, and these effects may be beneficial, harmful or neutral.

At the *population level*, mutations are the direct source of all new alleles and introduce genetic variation into a population. Mutations that are heritable can be passed on to future generations. If these new alleles are expressed as differences in phenotype, natural selection can act on these differences, so that undesirable mutations are removed from a population and desirable traits flourish. This in turn helps ensure the continuity of alleles that increase the chances of survival of organisms within the population – this is the basis of evolution by natural selection and speciation.

Worksheet
How the term 'mutation' was introduced into Biology

Weblink
Genetic variation: the outcome of mutation
Learn more by exploring mutations such as those in Shar-Pei dogs, red hair, cystic fibrosis and extra-toed cats.

Somatic and germline mutations – are all mutations inherited?

The type of cell in which a mutation occurs determines whether or not it will be inherited and, therefore, the impact of its effect. Most mutations arise in **somatic cells** (non-reproductive body cells) and are not inherited by descendants of that individual. However, some mutations occur in **germline cells** in the gonads (which produce gametes – egg and sperm cells). These mutations may be inherited by successive generations (Fig. 7.12).

Somatic mutations

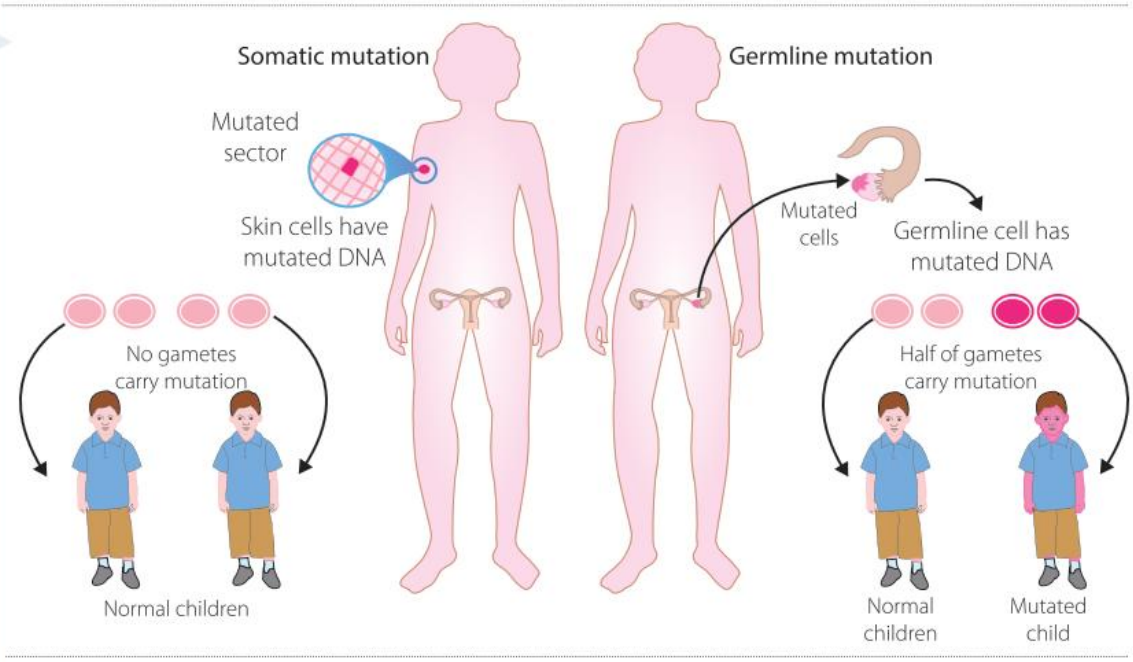
Somatic mutations occur in somatic cells, often due to replication errors prior to mitosis. Spontaneous mutations may occur in the S phase of the cell cycle (when DNA is vulnerable during replication) and, if not repaired during 'proofreading' in the G2 phase, will be passed on to daughter cells. When a mutated cell continues to divide by mitosis, the error is replicated each time and passed on to cells in successive divisions, amplifying the error within that tissue. This may result in an observable phenotypic difference in the tissue, such as pigmented cells in a carcinoma, a tumour or some other form of disease that is observable in an organ. Cancer is a common result of mutation in somatic cells – skin cancer, liver cancer and brain tumours are all examples of somatic mutations.

Somatic mutations do not always result in visible phenotypic changes in cells. Many occur as physiological changes, such as the mutations for cystic fibrosis, thalassaemia and Tay-Sachs disease. (Some of these diseases are discussed in more detail in Chapter 15.) The earlier in development the mutation occurs, the greater its effect will be, as the mutation will be replicated in a greater number of cells as the organism grows.

Germline mutations

Germline mutations (sometimes called **gametic mutations**) occur in the sexual reproductive cells that give rise to **gametes** (germline cells) and these mutations are passed to offspring. When a gamete carrying the mutation fuses with another gamete and an embryo forms, the mutation is replicated in every cell of the embryo as it divides and grows, affecting all cells in the resulting child.

FIGURE 7.12
Germline and somatic mutations



Coding and non-coding DNA

Review non-coding DNA in Chapter 4.

As you read in Chapter 4, some DNA codes for proteins (**coding DNA**) and some does not (**non-coding DNA**). The phenotype of an individual depends on the presence of particular proteins and the activity of those proteins. Mutations in coding genes therefore have a direct effect on these protein products. Mutations in non-coding DNA were originally thought to have no phenotypic effect, but it has been found that many affect gene expression, while some have no effect.

Mutations in coding DNA and their significance

In general, mutations in coding genes usually affect the type or sequence of amino acids in a protein end-product. In eukaryotes, mutations may also affect gene splicing and in this way modify the function or levels of the protein product. For example, whether a mouse's coat is white or grey depends on the presence of proteins, including enzymes that make the pigment. Because proteins are the product of genes in the coding region of DNA, if a mutation occurs in this region, it directly affects the protein and therefore the phenotype of the individual.

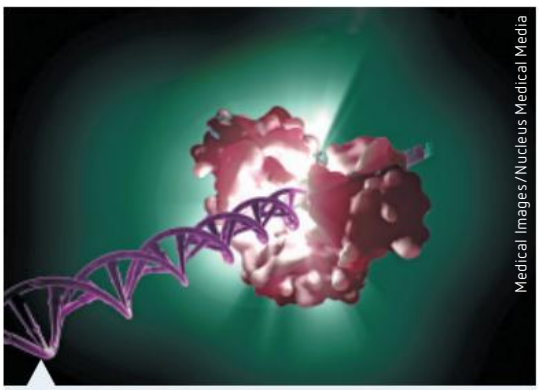


FIGURE 7.13 DNA repair enzymes wrap around a DNA molecule to repair it. Mutations in DNA repair enzymes can cause cells to malfunction, become cancerous or die.

Most DNA in prokaryotes is coding DNA; they do not have much non-coding DNA compared with eukaryotes. Studies of coding DNA in simple organisms such as bacteria and yeast reveal that it is, to a large extent, made of genes for DNA repair enzymes (Fig. 7.13). Maintaining the integrity of DNA is essential for the survival of these species. Microbes are exposed to constantly changing environments, but characteristics for DNA repair enzymes are not tolerant of genetic variation and change. In experiments where genes in coding DNA of prokaryotes were purposely inactivated, organisms showed an increase in the rate of mutation – both spontaneous and induced.

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Errors in the genes of eukaryotes whose products are involved in DNA repair are also serious, as the result is a general increase in mutations arising from errors in replication. An example of a mutation in a gene for DNA repair is a mutated gene that causes the disease *Xeroderma pigmentosum*. People who suffer from this disease are extremely susceptible to cell damage if exposed to UV light, and are a thousand times more susceptible to skin cancer than people who do not have the XP mutation.

Mutations in tumour suppressor genes can result in cancer, as can mutations in genes that activate proto-oncogenes (which normally promote cell division or reduce cell death).

Mutations in non-coding DNA and their significance

Mutations in the protein-coding regions of genes have commonly been associated with human genetic disease, but for many years molecular biologists were unsure of the function of non-coding DNA. In the 1950s, scientists discovered that some parts of non-coding DNA in eukaryotes have important functions, despite the fact that no protein end-product is made. They discovered regulatory sequences that promote ('switch on') genes or shut down ('switch off') genes. Many of these parts of non-coding DNA code for end-products other than DNA, such as rRNA and small nuclear RNA. These both have important functions in gene expression – small nuclear RNA plays a role in determining which introns are spliced out of DNA before it leaves the nucleus and rRNA is the machinery used by the cell for translation. Mutations in these non-coding regions have been shown to affect gene expression and cell functioning.

Research also showed that if segments of non-coding DNA were removed from an egg cell, it could not develop properly. There is growing evidence that mutations in non-coding genes are linked to developmental and congenital abnormalities (birth defects), supporting the idea that non-coding DNA is important during embryonic development. Mutations that occur in non-coding DNA in germline cells may lead to developmental abnormalities that cause the embryo or foetus to be aborted or, if less severe, may lead to congenital abnormalities. For example, an isolated (non-syndrome) congenital heart defect results from a mutation of the TBX5 enhancer gene in a non-coding area of DNA.

Studies involving genetic markers show that mutations in non-coding DNA have also been associated with a predisposition to specific diseases in adults. Mutations in the regulatory part of DNA, such as enhancer sequences, have been shown to genetically predispose people to non-infectious disease such as heart disease, diabetes, cancer and obesity, among other disorders. Some mutations in non-coding DNA are even associated with a predisposition to infectious disease such as hepatitis C. Scientists suggest that mutations in gene-regulatory DNA sequences may disrupt gene expression, leading to abnormalities in phenotype.

Evolutionists have suggested that non-coding DNA may hold an evolutionary advantage for the organisms in which it occurs. It may act as a buffer area dividing one gene from the next, so that if a frameshift mutation arises, the introns (gaps) will minimise the changes caused by the mutation. It may also play a role in giving some leeway when genes break during crossing over, so the recombination does not have to have pinpoint accuracy.

Junk DNA

Parts of non-coding DNA that seemed to have neither a protein-coding nor a regulatory function were given the name 'junk DNA' in 1972 by scientist Susumu Ohno. This term does not include regulating sequences in non-coding DNA (promoters, silencers and enhancers). Molecular biologists have also excluded from the 'junk DNA' category other non-coding DNA where specific functions have been identified (for example, DNA associated with the formation of centromeres and telomeres).

Only DNA that is highly repetitive is accepted by some molecular biologists as being of unknown function and termed 'junk DNA', because its nature and function remain a mystery.

More information on non-coding DNA is provided in Chapter 4, page 124.

Worksheet
The biochemical nature of mutations

This portion of non-coding DNA contains hundreds and thousands of repeats of DNA sequences that are just a few hundred base pairs long. The origins of these repeat sequences are believed to be viruses that inserted copies of a segment of their genetic material into a host cell's DNA. Inserted DNA elements are called 'transposable' elements and may be **transposons** (transposed DNA) or **retrotransposons** (inserted viral RNA that has been reverse-transcribed back into DNA). Transposons and retrotransposons are considered mutations if they occur near genes or in their promoter regions, where they change gene expression or functioning.

The large numbers of repeat DNA sequences are believed to have arisen when these elements made numerous copies of themselves within the DNA of the cell. Some geneticists call these repeats 'selfish DNA', as it uses host cell material and gives nothing back. This raises the question of why they have not been removed from the genome if they serve no function. Some scientists argue that, from an evolutionary perspective, transposable elements introduce variation and keep the genome diverse, providing increased opportunity for adaptation by natural selection. The debate among scientists continues.

INVESTIGATION 7.2

Analysing data to investigate mutations in coding and non-coding DNA

Current projects like ENCODE and the Epigenomics Roadmap are researching the 98% of the genome that comprises non-coding DNA, to determine whether it is as essential to gene function as coding regions.

In this investigation, you will investigate the relationship (if any) between quantity of DNA and how complex an organism is, and you will analyse data to assess the significance of 'coding' and 'non-coding' DNA segments of DNA in the process of mutation.

Information and communication technology capability

Numeracy

Critical and creative thinking

PART A

Early geneticists thought that the amount of DNA in the genome of an organism would be an indication of its complexity.

METHOD

Investigate the validity of the hypothesis proposed: that the amount of DNA in a genome correlates with the complexity of the organism. Find data in the form of tables and graphs to support your answer. Write a conclusion for your findings and write a discussion paragraph, explaining the science behind your conclusion.

RESULTS

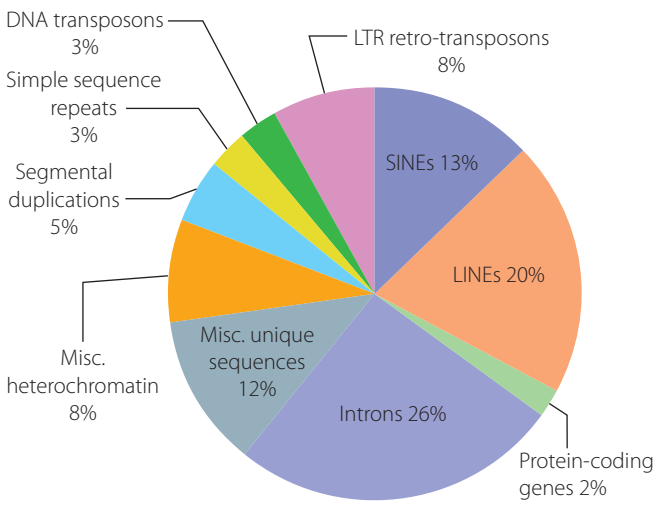
1 Write a conclusion about the validity of the hypothesis.

2 Provide evidence to support your conclusion (text, tables and graphs).

PART B

METHOD

1 Analyse the pie graph in Figure 7.14 and answer the questions that follow.



LTR = long terminal repeat
SINEs and LINEs are types of retrotransposons.
Heterochromatin is known to play a role in gene expression.

FIGURE 7.14 Components of the human genome

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» 2 What proportion of the human genome is made up of:
   a protein-coding genes
   b non-protein-coding genes
   c transposable elements
   d retrotransposons
   e unique sequences with no known function
   f duplications and repeats?

3 From the pie chart, calculate the proportion of non-coding DNA that you would consider to:
   a be junk DNA. Justify your answer.
   b play a role in gene expression.

4 Write a research-based answer to the syllabus outcome: *Assess the significance of 'coding' and 'non-coding' DNA segments in the process of mutation*. Your answer should be approximately 200–300 words.

   Before tackling this question, use information from this section of the textbook, as well as from your answers to Questions 1 and 2, to write some research questions that outline the information you need to find out. Use secondary sources to gather information to address your research questions.

DISCUSSION AND CONCLUSION

Use information from both your research and from the graph in Figure 7.14 to support your answer to the inquiry question at the start of this chapter: How does mutation introduce new alleles into a population?

Investigating causes of genetic variation: fertilisation, meiosis and mutation

Mechanisms of sexual reproduction such as gamete formation (meiosis) and fertilisation increase gene recombination and therefore variation in individuals (new gene combinations) and variability in a population (the amount by which individuals in a population vary from each other genetically). Evolutionary studies show that greater variability improves the ability of a population to adapt to changes in the environment, resulting in an increased chance of survival. If there is little or no variability within a population, the result is a static or unchanging population that is less likely to be able to adapt to sudden changes in the environment, and more likely to be wiped out.

As a very simple example to illustrate variability in a population, consider humans. One parent may have blue eyes and fair hair, the other dark eyes and dark hair. If the traits for hair colour and eye colour assort independently of each other – that is, they occur on separate chromosomes – their gametes may combine to produce offspring that have blue eyes and dark hair, or brown eyes and fair hair. In these cases, the offspring have a different combination of genes than their parents, increasing variability. Variability in a population may be further increased if there are a *greater number of alleles* present for each gene. If, within the human population under discussion, there are also individuals with red hair and green eyes, there is greater variability and an even greater opportunity for more gene combinations to arise in gametes produced by individuals.

Therefore variability may be increased as a result of:

- recombination of genetic material
- an increased number of alleles for a particular gene.

Mutation plays a part in increasing the number of alleles for a trait, whereas meiosis and fertilisation (gamete formation and gamete fusion) play a significant role in recombining genetic material.

Remember, although the basis of inherited variation is mutation, not all variation – and therefore variability – is genetic in origin. The effect of the *environment* on variation (phenotype) of organisms was discussed in Chapter 4.

Meiosis and genetic variation

When gametes form, *crossing over* and *random segregation* in meiosis result in **genetic recombination** of paternal and maternal genes within each gamete. When two different gametes unite during sexual reproduction (*fertilisation*), a further mixing of genetic material occurs and this results in offspring with many new combinations of genes. The importance of this variability is described below.

Variation during meiosis and gamete formation may also be caused by *mutations* – either during replication of DNA (leading to replication errors) or during the separation (disjunction) of chromosomes (leading to chromosomal mutations). Replication errors in meiosis may arise in a similar manner to those in mitosis, commonly leading to point mutations. Chromosomal mutations may arise due to errors in crossing over or non-disjunction of chromosomes or chromatids during anaphase I or II.

Chromosomal errors

When crossing over goes wrong, chromosomal aberrations may be introduced. For example:

- the DNA to be exchanged during synapsis may be inverted before it is inserted onto the arm of a corresponding chromatid
- a chromosome may break and, if this is followed by a duplication of the chromosome, the two chromatids that fuse may now have two centromeres. When they pull apart in anaphase, one chromatid will have a duplication and one will have a deletion.

These chromosomal changes may be brought about by exposure to mutagens during meiosis. Remember that gametes may remain in meiosis I for a prolonged period of time in females (many years, because the gametes are formed in embryonic life) and so exposure of female ovaries to mutagens at any stage during reproductive life may be detrimental. This is why females are given an X-ray-deflecting apron to wear when having X-rays.

Weblink
What happens when mitosis goes wrong?
Changes in chromosome number and structure, mitotic errors and cancer.

Changes in chromosomal numbers (non-disjunction)

When one or more pairs of homologous chromosomes or sister chromatids do not separate as they should during nuclear division, an abnormal distribution of daughter chromosomes in the resulting daughter cells occurs, and this may lead to a change in chromosome number (Fig. 7.15). An example of this is Down syndrome, caused by an extra copy of chromosome 21. The age of the parents, as well as some environmental factors, may increase the risk of errors in meiosis leading to chromosomal mutations.

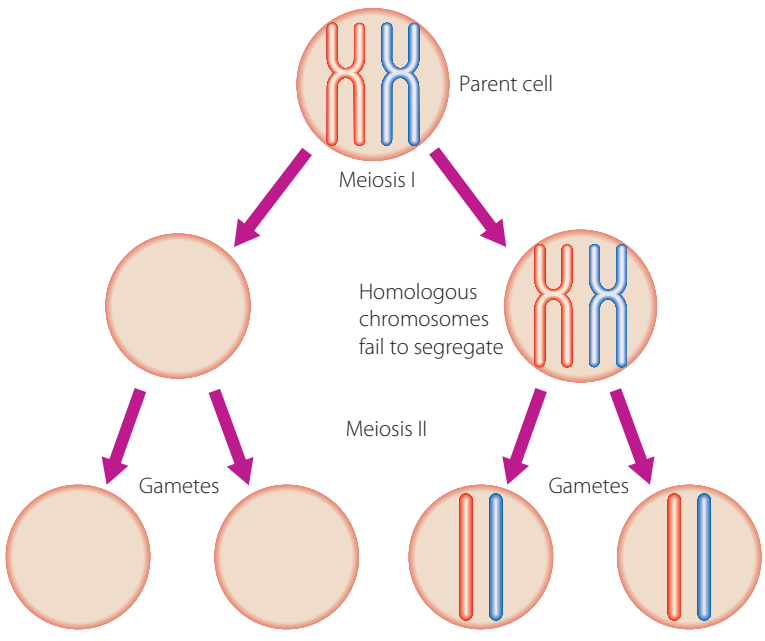

FIGURE 7.15 Non-disjunction of chromosomes during meiosis

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INVESTIGATION 7.3

A secondary-source investigation of the causes of mutation related to meiosis and fertilisation

In non-disjunction, a pair of chromosomes fails to segregate during meiosis. Therefore one resulting cell (gamete) may inherit the pair of chromosomes, while the other inherits neither chromosome from that bivalent. Each of the resulting gametes will have an incorrect chromosome number.

See Chapter 15 for examples of disorders arising as a result of non-disjunction.

METHOD

Use a range of secondary sources to investigate the effect of the age of parents (mother and/or father) and the impact of one environmental factor (such as alcohol or smoking) on the rate of mutation during gamete formation and fertilisation.

Support your findings with quantitative data.

Numeracy

RESULTS

Present your information in the form of a brochure to educate the public.

CONCLUSION

Write a statement on your brochure, beneath the data, summarising your findings.

KEY CONCEPTS

- Mutations cause diversity in populations by introducing genetic variation.
- In multicellular organisms, mutations can be classified according to the type of cell in which they occur.
  - Somatic mutations arise in a single body cell and cannot be inherited. They affect only patches of tissue derived from the mutated cell and are more severe if they occur early in development.
  - Germline mutations arise in gametes and can be inherited, affecting all cells in the individual that the gamete gives rise to.
- Variation is introduced during meiosis when genetic recombination occurs during crossing over and random segregation.
- When two different gametes fuse during fertilisation, a further mixing of genetic material results in offspring with many new combinations of genes.
- Another cause of variation during meiosis and gamete formation may be mutations arising during replication of DNA or during the separation (disjunction) of chromosomes.
- Mutations and sexual reproduction are the reason that organisms are not genetically identical, even if they have the same ancestors.

CHECK YOUR UNDERSTANDING 7.3

1. Define 'mutation', 'variation' and 'variability'.
2. Compare germline mutations and somatic mutations.
3. Distinguish between coding and non-coding DNA.
4. What is the function of coding DNA?
5. List three known functions of non-coding DNA.
6. Outline the effects of mutations that occur in coding DNA and explain how these are different from the effects of mutations that occur in non-coding DNA.
7. Give one example of a disease that arises as a result of non-disjunction of chromosomes in meiosis.
8. Compare how chromosomal aberrations and changes in chromosome numbers arise during meiosis.
9. Explain three ways in which genetic variation may be introduced during meiosis and fertilisation.

7.4 Population genetics – mutation, gene flow and genetic drift

The genetic diversity in plant and animal populations today is the result of global spread and each population adapting to its local environment. New alleles may be introduced into a population by mutation, or through the movement of individuals between populations (emigration and immigration). Genetic diversity is the result of a large number of variants (different alleles) that may be present for each gene and a large number of genes that may be present for each trait (polygenic traits).

For example, in the human population, the gene for height is not like Mendel's pea plants where there are only two alleles – tall or short. In humans, height is a polygenic trait and each gene for the trait has two or more alleles. As a result, there are a large number of variants – one polygenic trait (height) has many possible genotypes and phenotypes.

In a graph showing the height of individuals against how many in the population have each height (frequency of phenotype), the curve has a continuous distribution. If you measure the height of all students in your class and calculate an average, you will find that many students are a little taller or shorter than the average, with a few students much taller or much shorter if the sample size of your class is large enough. On a graph, this distribution appears as a bell-shaped curve, termed a 'normal distribution curve', typical of polygenic traits (Fig. 7.16).

As you know, one of the main sources of genetic variations is mutation. This affects phenotype, the basis for natural selection. The graph in Figure 7.17 shows the effect of mutations on the fitness of individuals in a population. Very few mutations overall are advantageous and selected to increase in frequency. Detrimental or deleterious mutations (highest frequency) are eliminated, neutral mutations remain and those few that are beneficial are selected. Neutral mutations that are maintained in the population can be considered an evolutionary 'back-up' – they provide variations that have no immediate effect, but may provide an advantage in the future if the environment changes suddenly.

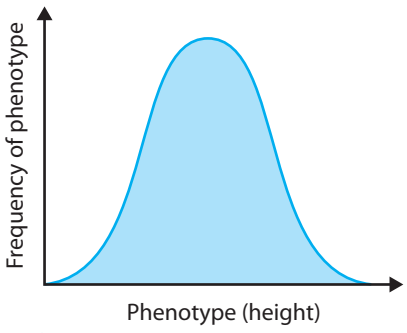

FIGURE 7.16 The distribution of phenotypes that would be expected for a polygenic trait such as height, where many genes contribute to the trait

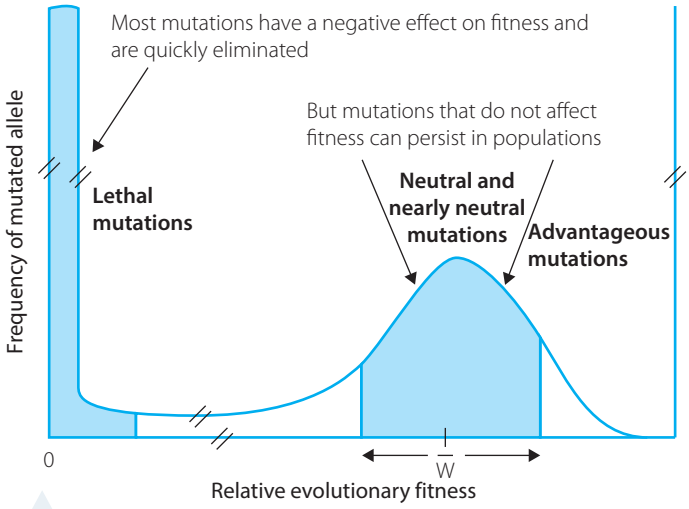

FIGURE 7.17 A hypothetical distribution curve showing the relationship between new mutant alleles and natural selection. *Relative evolutionary fitness* is a measure of the survival and/or reproductive rate of a genotype or phenotype. W is the average range of fitness for an allele in the population.

Population genetics and factors causing changes in allele frequency

Population genetics is the study of how a population changes over time, leading to species evolving. Population genetics involves *quantitative* study (looking at quantitative data – numbers) in which

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scientists analyse the *distribution of genetic variation* in a population. To do this, population geneticists study *factors that cause increases or decreases in allele frequency* within a population.

Natural selection is not the only factor that causes changes in the distribution of genetic variation in a population, resulting in it changing over time. Sometimes individuals in a population are not selected by natural selection, but instead survive and reproduce by chance. For example, a natural disaster may strike in a particular place and wipe out some organisms, while others have a lucky escape. The allele frequency of the survivors will increase as they reproduce. This change in allele frequency due to chance is known as **genetic drift**. Another way in which allele frequency may change is due to new individuals entering a population, or existing individuals leaving (**gene flow**).

Sometimes an allele may become 'fixed' in a population. This means that it becomes the only remaining allele in that population, having outcompeted all other less successful alleles. This is very unusual, because most variants or alleles only give a small benefit or no benefit at all.

Therefore, although the likelihood that a variation (mutation) will be passed on to subsequent generations is often due to choice (natural selection), it may also be a result of chance (genetic drift) or immigration and emigration (gene flow).

A population is a group of individuals of a species that live in a common area and are interbreeding. Allele frequency is a measure of how often an allele occurs in a population.

Weblink
Gene flow and genetic drift – a simulation
Work through the simulation to increase your understanding of gene flow.

Scientists who paved the way for population genetics

To study population genetics, you need to use your knowledge and understanding of the laws of *genetic inheritance* to predict how the genetic composition of a population will change when subjected to *evolutionary selective pressures*.

Progress in the field of population genetics depends on the work of many scientists. Mendel and Darwin paved the way, and other biologists have worked on developing mathematical models that can be used to make predictions for current populations. These mathematical models take Mendel's and Darwin's findings into account.

- Mendel's findings:
  - Each parent donates one allele for every gene to their offspring; therefore, offspring have two alleles for every gene.
  - Some alleles are dominant and expressed, whether they are present in a single copy or two copies in an individual.
  - Other alleles are recessive and only expressed when they are not paired with a dominant gene.
- Darwin's findings:
  - Natural selection is the primary driving force for evolution; if a gene confers an advantage, it is more likely to be passed on to the next generation.

Factors that cause changes in allele frequency within a population

1. *Selective pressure* causes changes in allele frequency due to variations – alleles that make individuals well suited to the environment increase.
2. *Sexual selection* (or non-random mating) changes allele frequency because mating is not random; the most successful maters' genes remain in the gene pool.
3. *Mutation* leads to the formation of new alleles, due to changes or 'errors' in DNA that arise during gametogenesis (meiosis). During fertilisation, 'good' (useful) and/or 'bad' (deleterious) errors are passed on to the next generation.
4. *Genetic drift* causes changes in allele frequency due to random chance. This causes individuals to be different, not necessarily more successful. Genetic drift may occur as a result of a natural disaster (**bottleneck effect**) or due to a few individuals in a population (founding individuals) becoming geographically isolated from the original population (**founder effect**). The remaining individuals may not be an accurate representation of the allele distribution and genotype frequencies of the original population. The change in allele frequency due to genetic drift is greater if the population is small.

Weblink
Virtual biology lab
Population genetics simulations

5 *Gene flow* changes allele frequency due to mixing of new individuals in a population. As a result, different alleles find their way into the population (for example, through immigration) and these genes spread. Alleles may be lost from the population through emigration. This effect is also more noticeable in smaller populations.

Today, technology and genetic testing, together with an understanding of heredity, allow us to study populations over several generations and observe evolution happening (rather than having to observe them over millions of years). Population geneticists are able to use this data to describe how genetics and evolution influence each other, using mathematical models they have created.

Although all humans are genetically very similar, no humans (other than identical twins at birth) are genetically identical. In terms of DNA sequences, all humans are 99.9% similar to each other in genetic composition. Our differences are an indication of our genetic variation. We share 99% of our genes with chimpanzees and 60% with chickens. However, even a 0.1% difference in the DNA of individual people still amounts to hundreds of thousands of base pairs. The human genome consists of approximately 3×10^9 (3 billion) base pairs, of which 6×10^6 (6 million) are different. As you learned in the previous chapter, analysis of SNPs (single nucleotide polymorphisms) may be used, rather than looking at actual DNA base sequences, to take into account these differences in human DNA. SNP studies are important in enabling scientists to search for variations in DNA associated with particular human traits, including susceptibility to disease and response to certain medical drugs, which in turn may affect their treatment.

We know that *genetic stability* occurs if all individuals have the same reproductive capacity and fitness. Any change that causes some individuals to produce more offspring than others, such as an increase in an individual's ability to survive, mate and reproduce, will result in a change in allele frequencies. In a similar way, if an allele makes an individual more vulnerable to disease or predation, it is likely to decrease in frequency, until it is either very rare or eliminated from the population.

All of this must be taken into account when developing models to predict changes that may occur in allele frequency in populations and identify causes of these changes.

INVESTIGATION 7.4

Secondary-source investigation to evaluate the effect of mutation, gene flow and genetic drift on the gene pool of populations

BACKGROUND

Some diseases that have arisen as a result of mutation occur in different frequencies in the gene pools of various populations in the world today. Exploring the reasons for the changes in the frequency of these diseases is interesting. For some diseases, the effects of the mutation may be twofold – detrimental in one respect, but conferring an advantage to certain populations in another. The frequency of some diseases is not affected by geographical distribution, but may be affected by the age of individuals within the population. Alleles of other diseases are not reduced because their disadvantageous symptoms only appear once an individual is past reproductive age.

In this investigation, you will look at the average frequency of a given disease in the human race and compare this with the frequency in different population groups, investigating a possible cause for this change in frequency. At the end of the investigation, you will evaluate the effect of mutation, gene flow and genetic drift on the gene pool of these populations.

The parts of this investigation differ in complexity. All students should be able to complete parts A and B. Part C is for advanced students or those who wish to extend their knowledge further.

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Part A (Basic investigation – all students)

AIM

To investigate the impact of mutation, genetic drift, gene flow and natural selection on a hypothetical population of stickleback fish

METHOD

Work through the interactive video simulation on the weblink and identify, in the evolution story of these sticklebacks, when and how genetic drift, gene flow and natural selection occurred.

Weblink
Population genetics – sticklebacks

RESULTS

Write a hypothesis, method and results for the experiment described in the simulation. Copy and paste the graph showing % frequency of sticklebacks over time. Calculate the percentage change in frequency of the two genotypes of sticklebacks within the population in 1983, 1993 and 2003.

Numeracy

CONCLUSION

1. Write a statement summarising the findings of the experiment.
2. Evaluate the effect of mutation, gene flow and genetic drift on the gene pool of the stickleback population.

Part B (Intermediate level)

AIMS

To carry out a case study of a genetic disease caused by different types of mutations and investigate:

1. the cause of the genetic variation (mutation, mutagen, process – meiosis/fertilisation)
2. the frequency of the disease in populations around the world
3. the effect of gene flow and genetic drift on the distribution of the disease
4. the correlation (if any), in big data sets, between the incidence of the disease and the number of Internet searches for information about the disease, around the world

Information and communication technology capability

METHOD

1. Conduct a case study of three diseases, each caused by one of the following types of mutation:
   - **A** point mutation – for example, sickle cell anaemia
   - **B** chromosomal mutation – for example, Huntington's chorea or fragile X syndrome
   - **C** change in chromosome number – for example, Down syndrome.
2. Recommended procedure: work in general groups of six, carrying out a jigsaw activity, two researching disease A, two researching B and two researching C.

   These expert pairs from each general group may join up to form an expert group to investigate the disease they have been allocated. Each expert pair will report back to their original general group.
3. In expert groups or pairs, begin your research using the weblinks provided and then go on to use a variety of sources. Research the information required for your particular case study, to meet the requirements outlined in the Results section.
4. The general group re-forms, with all six students coming together to collate their information on the three different types of disease, reading each other's information and answering the three to five questions posed by each expert pair. (See Results.)

Weblink
Genetic diseases
Sickle cell anaemia
Huntington's chorea
Down syndrome
Fragile X syndrome

RESULTS

Present your results (in a collaborative way, such as Google Docs), including the following information and data for each disease that has been investigated:

1. Type of mutation (and draw a diagram to illustrate)
2. Number of variants (one or more mutations) responsible for causing the disease in populations
3. Possible mutagens (and reasons why you think these may be the cause of the mutation)

Refer back to your findings in Investigation 7.1 to help you with point 3 of the results.

» 4 Process by which the mutation arose – when, how and where. For example:
- when – during replication/in mitosis/non-disjunction in meiosis, and so on
- how – describe the process
- where – whether you think this occurred in a germline or somatic cell mutation (and reasons for your choice).

5 Average frequency of the disease in all humans in the world, compared with frequencies in different populations on continents/in countries around the globe. Include a *map* to illustrate the frequency of the disease in different parts of the world, as well as a *graph* showing relevant data. Investigate possible reasons for this distribution (include the impacts of gene flow, genetic drift and natural selection).

Information and communication technology capability

6 Use the 'Google explore data' weblink and prevalence data to find out whether there is a correlation between the number of Google searches for this type of information and the part of the world where alleles for the disease occur in the highest frequency (that is, where the disease is most prevalent). Write a paragraph to outline your findings. Use data to justify your conclusion.

Weblink
Google explore data
Do a correlation study of the number of Google searches for this type of information and compare it with your findings on frequency of disease in Question 5.

CONCLUSION

Evaluate the effect of mutation, gene flow and genetic drift on the gene pool of the population with the highest incidence of this disease.

DISCUSSION

Write 3–5 questions based on your findings, for your peers to answer. Each question should be of a different level of complexity, ranging from 1–5 on a scale, where:
1 = Recall of a small amount of knowledge and understanding is required to answer the question.
5 = A large amount of knowledge and understanding is required, and skills in analysing and synthesising information are necessary to answer the question.

Part C (Intermediate/Advanced – extension)

Read the information about the Hardy-Weinberg principle and work through the simulations on the weblinks that follow.

Worksheet
Introducing the Hardy-Weinberg equation

The **Hardy–Weinberg principle** (also known as the Hardy-Weinberg equilibrium) is based on the idea that the frequency of alleles in a population remains constant from one generation to the next if none of the evolutionary influences (such as mutation, sexual selection, genetic drift or gene flow) are acting. That is, it assumes that a population is in a hypothetical state of equilibrium.

G.H. Hardy and William Weinberg introduced their mathematical model in 1908 and developed an equation that shows how Mendelian genetics work at the level of the whole population and gives the expected frequency of different alleles in a hypothetical population that is not evolving.

It makes certain assumptions about the hypothetical population:

- alleles are equally beneficial (no natural selection)
- mating is completely random (no sexual selection)
- no random modification of the gene pool occurs (no mutation)
- the population is large (no effect from genetic drift)
- no immigration or emigration occurs (no gene flow).

Weblink
Basic/intermediate/advanced: Population genetics
Use different models to explore evolutionary influences on the frequency of alleles and genotypes in virtual populations.

The Hardy-Weinberg principle enables scientists to find a relationship between the phenotype and the actual frequency of the genes in a population. If we know the frequency of homozygous recessive individuals in a population (for example, the percentage of the population that is ww), we can conclude that the remaining individuals must be either WW or Ww. We can then use simple mathematics to work out what proportion of the remaining population is WW and what proportion is Ww.

We can then look at actual allele frequencies and try to find reasons for the difference between this and the expected ratios.

The change in allele frequencies in a gene pool of a population over generations is known as microevolution. Populations may undergo microevolutionary change and still interbreed, but at times isolation occurs and this may lead to the development of new species (speciation). This means that if the original populations were brought together again, they would have enough variation that they could no longer interbreed, indicating that, over many generations, microevolutionary changes can lead to new species. Therefore the Hardy-Weinberg equation can be a useful way to describe the degree of microevolution that is taking place over successive generations.

Weblink
Intermediate/advanced extension: Deriving the Hardy-Weinberg equation

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KEY CONCEPTS

FACTOR	WHAT IS IT	CHANGE IN ALLELES IS DUE TO ...	EFFECT ON THE NEXT GENERATION
Selective pressure	The main selective pressure is natural selection (Darwin)	variations that are passed on because they make individuals more likely to survive (and more virile)	Alleles that make individuals 'fitter' – more likely to survive and live to reproductive age – become most frequent
Sexual selection	Certain individuals are more attractive to mates and therefore more likely to breed	non-random mating (mating is not random; some individuals mate more than others)	Alleles of individuals who are most successful at mating are more common in the gene pool
Mutation	New genes arise due to 'errors' in DNA replication during meiosis (gametogenesis); they may be beneficial, neutral or harmful	new alleles arising during gametogenesis being introduced into a population	New alleles that are beneficial become more frequent in the population
Genetic drift (more obvious in smaller populations)	Random events (e.g. a tornado) lead to a change in gene frequency because some individuals are wiped out	random chance (non-selective; does not depend on genetic make-up)	Causes individuals within a population to be different (not necessarily more successful) due to random chance
Gene flow (more obvious in smaller populations)	Individuals with different genes come into a population and spread their alleles	mixing with new genetically different individuals (e.g. immigration, emigration)	Allele frequency in the population changes

CHECK YOUR UNDERSTANDING 7.4

1 Match the following terms with the statements below.

genetic drift, gene pool, genome, gene flow

- **a** Individuals survive and reproduce due to chance, resulting in a change in allele frequency.
- **b** New individuals enter or leave a population.
- **c** The range of genes and all their alleles in a population.
- **d** All the genetic material in an individual or in a cell.

2 Explain the role of selective pressure in microevolution.

3 How do sexual selection, genetic drift and gene flow affect allele frequencies within a population?

4 Explain why genetic drift and gene flow show a greater effect on allele frequencies in smaller populations.

7 CHAPTER SUMMARY

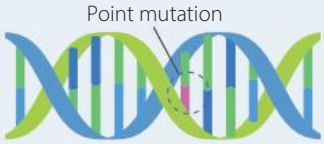
Mutation: What is a mutation?

DNA CHANGE

At the molecular level

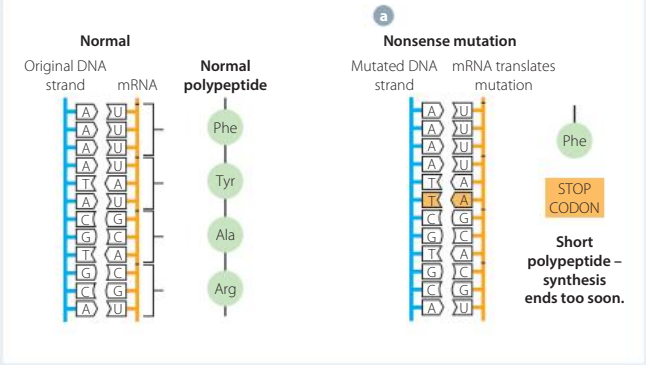
A change in nucleotide sequence

- point mutation



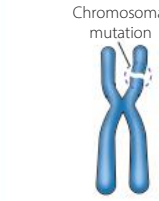
Types of point mutations

- Nonsense
- Missense
- Silent
- Frameshift insertion
- Frameshift deletion



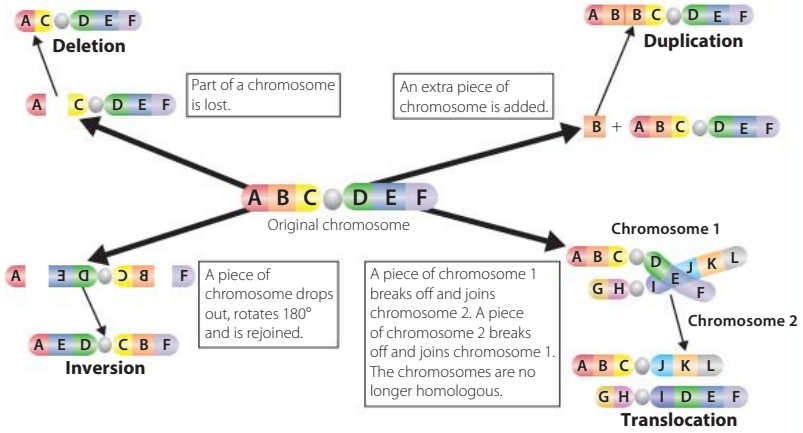
At the chromosomal level

(change in chromosome structure/number)

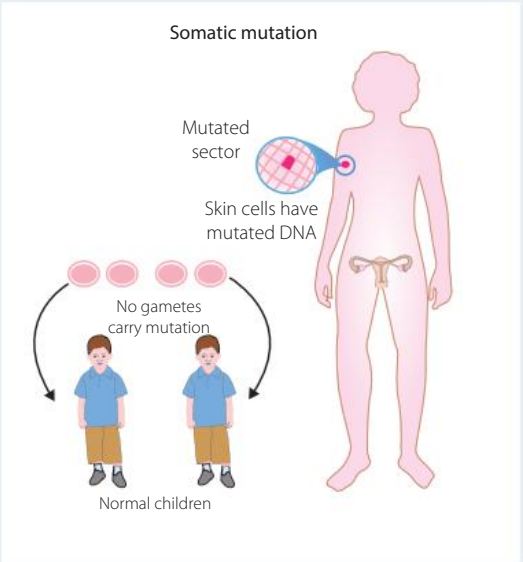


Chromosome structure mutations are the result of alterations to the structure of one or more whole chromosomes.

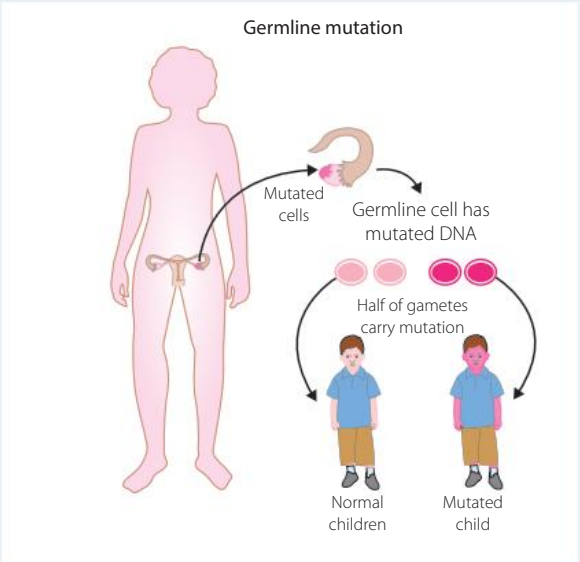
- Chromosomal mutations
- insertion
- deletion
- inversion
- duplication



At the cellular level



At organism level



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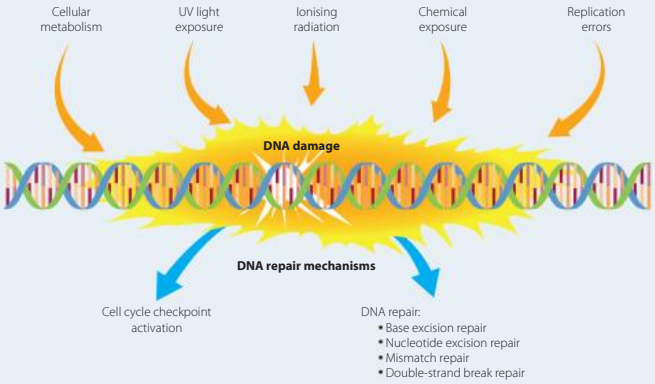
HOW ARE NEW ALLELES INTRODUCED?

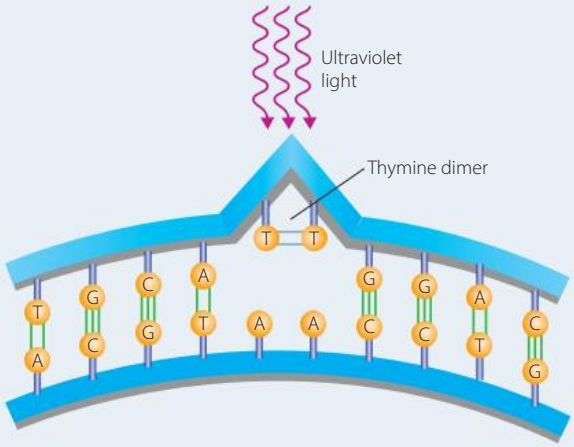
Mutagens

- Physical e.g. EM radiation
- Chemical e.g. tobacco smoke
- Naturally occurring e.g. viruses

Mutation and genetic variation

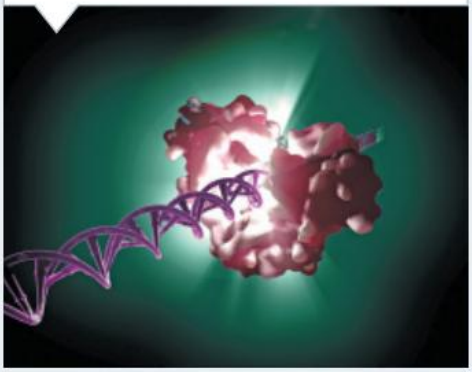
Mutagen → Mutation → Physical change in DNA → new allele





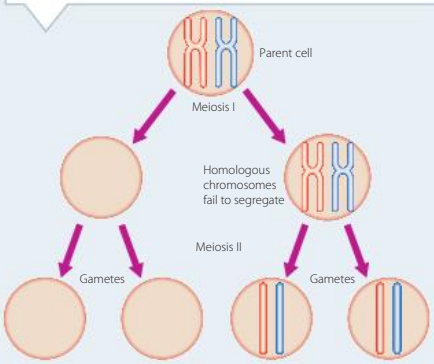
DNA repair

- Mutations can affect DNA repair

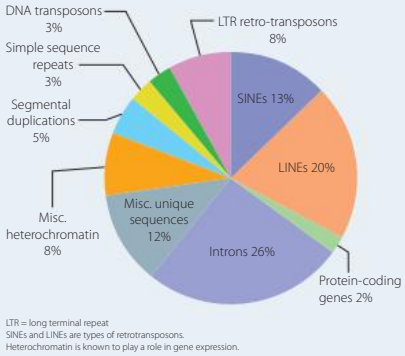


MEIOSIS

- non-disjunction

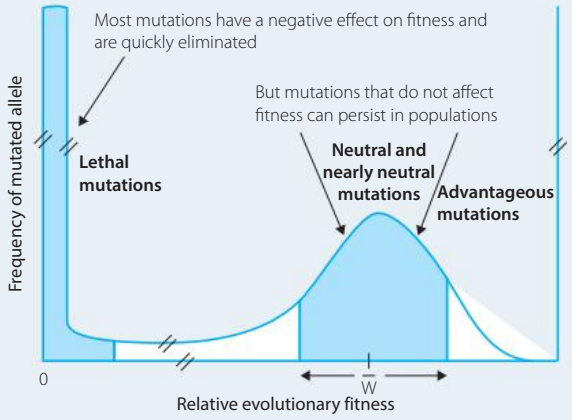


CODING/NON-CODING DNA



LTR = long terminal repeat
SINEs and LINEs are types of retrotransposons.
Heterochromatin is known to play a role in gene expression.

Mutations can lead to Changes in alleles at population level



7 CHAPTER REVIEW QUESTIONS

Review quiz

1 Distinguish between the following terms:
- **a** mutation and mutagen
- **b** spontaneous mutations and induced mutations
- **c** deletion and duplication
- **d** inversion and translocation
- **e** somatic mutation and germline mutation
- **f** missense mutation and nonsense mutation
- **g** aneuploidy and polyploidy
- **h** coding DNA and non-coding DNA
- **i** gene flow and genetic drift
- **j** point mutation and chromosomal mutation
- **k** polygenic traits and multi-allelic traits.

2 Explain what is meant by the following categories of mutagenic agents, and give two examples of each:
- **a** electromagnetic radiation
- **b** chemical mutagens
- **c** naturally occurring mutagens.

3 Copy and complete the table to summarise how mutagens operate in cells.

MUTAGEN	TYPE	EFFECT ON DNA	EXAMPLES
Chemical	Ingested chemicals		
	Environmental irritants and poisons		
Naturally occurring mutagens	Biological		
	Non-biological		
Electromagnetic radiation	Ultraviolet radiation		
	Ionising radiation		

4 Outline the processes during which mutations can arise.

5 Using words and arrows, construct a flow chart to show how a change in DNA can lead to a change in phenotype.

6 What is a point mutation? Give examples.

7 What is the difference between a chromosomal mutation and a gene mutation?

8 Explain, using letters to represent bases, each of the following types of gene mutations:
- **a** base substitution
- **b** frameshift.

9 Explain, using diagrams where necessary, the following types of chromosomal mutations:
- **a** deletion
- **b** duplication
- **c** inversion
- **d** translocation.

10 Explain how non-disjunction in meiosis can lead to trisomy. Use a diagram to help you explain.

11 Discuss the advantages and disadvantages of DNA having the ability to repair itself.

12 Describe the way transposable genetic elements operate, and discuss their impact on the genome.

13 Why do germline mutations exert a greater phenotypic effect than somatic mutations?

14 Why are somatic mutations not passed on to offspring?

15 Explain how fertilisation, meiosis and mutation lead to genetic variation.

16 Name five factors that cause changes in allele frequency within a population, and explain how each brings about this change.

17 Explain why the sickle cell mutation is described as a missense point mutation.

18 Draw a flow chart to illustrate a silent mutation and the resulting polypeptide chain that would be produced. (See Fig. 7.10 for ideas.)

19 Explain how mutations in DNA may lead to a generation of new alleles.

20 Explain, using examples, how DNA mutations may lead to cancer.

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21 Construct a table to summarise the types of point and chromosomal mutations, their causes, the changes in DNA structure (process of mutation) and the effects of each mutation. Draw a diagram to show an original base sequence and the resulting changed base sequence for each type of mutation. Use the following headings: Type of mutation, Causes, Change to DNA, Effect of mutation, Example of disorder, Diagram.

22 Using examples, evaluate the effect of each of the following on the gene pool of populations:

- **a** mutation
- **b** gene flow
- **c** genetic drift.

23 The change in allele frequencies in the gene pool of a population over generations is known as microevolution. Explain how mutation can lead to microevolution and speciation over time.

24 Using Figure 7.18, explain the difference between somatic and germline mutations when gametes are produced during meiosis.

25 Is it true to say that mutations in coding DNA have a phenotypic effect, but mutations in non-coding DNA have no phenotypic effect? Justify your answer.

26 In nature, mutation occurs spontaneously at a very low rate. As a result, geneticists at first found it difficult to investigate mutations and their effects. In 1927, American biologist H.J. Muller discovered that irradiating fruit flies (*Drosophila melanogaster*) with X-rays could greatly increase the mutation rate. Explain how this discovery could be applied to study the cause and inheritance of mutations.

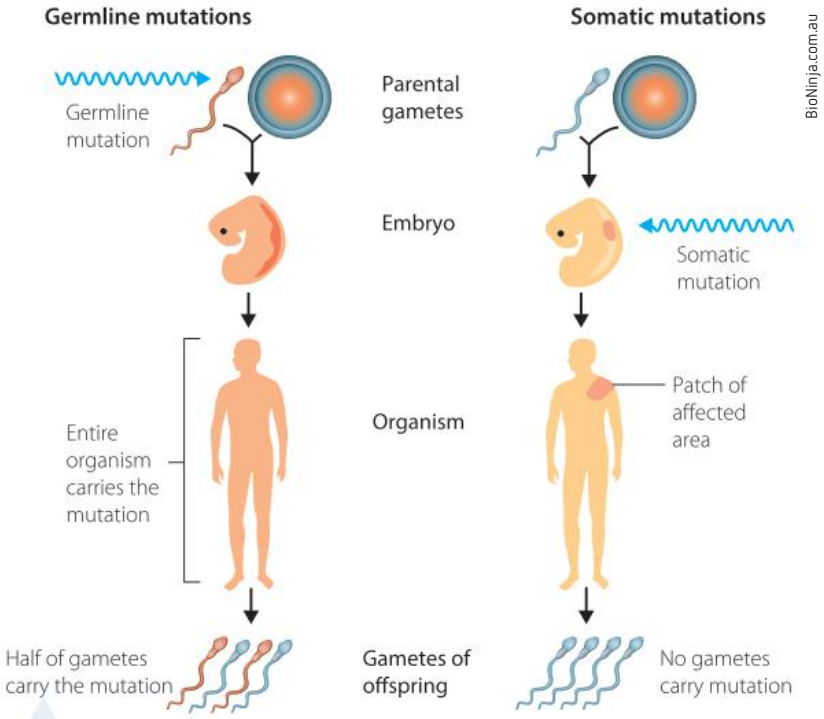

FIGURE 7.18 Results of germline and somatic mutation

Exam preparation

8 Biotechnology

INQUIRY QUESTION

How do genetic techniques affect Earth's biodiversity?

Students:

- Investigate the uses and applications of biotechnology (past, present and future), including: (ACSBL087)
  - analysing the social implications and ethical uses of biotechnology, including plant and animal examples S EU ICT CC
  - researching future directions of the use of biotechnology CCT ICT
  - evaluating the potential benefits for society of research using genetic technologies S EU PS
  - evaluating the changes to the Earth's biodiversity due to genetic techniques S EU PS

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
- **8.1** Secondary-source investigation into the uses of biotechnology: past, present and future
- **8.2** Secondary-source investigation to research an ethical issue
- **8.3** Secondary-source investigation of the future direction of biotechnology
- **8.4** How do genetic techniques affect Earth's biodiversity?

Worksheets
- Ancient biotechnologies
- Technologies to manipulate DNA
- Modern technologies
- Use of modern biotechnology in conserv White rhinos

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

The term 'biotechnology' is used in all spheres of life today – from areas such as the food industry, agriculture and conservation, to the health industry, medical circles and business. This topic of interest surfaces not only at global conferences, but in all forms of media, social networking, business meetings and even around dinner tables and in coffee shops. Discussion of biotechnology can evoke strong emotional responses, based on differing opinions. So what, exactly, is biotechnology?

Biotechnology literally means the use of biological materials (*bio*) as tools (*technology*). Simply put, it is the use of living organisms or their products to fulfill human needs. Common to the many formal definitions of biotechnology is the idea that living materials, biological processes or biological products are used to make new products that are useful to humans, in fields such as industry, agriculture and medicine. Examples of biological materials from living things include cell products (such as enzymes or antibiotics), living cells, tissues and whole organisms (micro-organisms, plants and animals). Although biotechnology has many beneficial applications, biotechnology used by humans may be detrimental to the environment or other humans, plants or animals. Taken to the extreme, some biotechnologies may be used for destructive purposes, such as in 'bioterrorism'. As a counter-measure, the field of bioethics has grown, so that people can put in place boundaries by looking at the social and ethical impacts of biotechnology, its uses and its potential further applications.

FIGURE 8.1 Biotechnology – past and present: **a** discovering penicillin; **b** producing a DNA micro-array

Biotechnology includes ancient and traditional biotechnologies used to make beverages such as wine and beer, and foods such as bread, yoghurt and cheese. It encompasses processes in agriculture to manipulate plant and animal breeding to produce higher milk-yielding animals and disease-resistant crops, as well as modern genetic technologies involving gene manipulation. It also has ground-breaking applications in medicine and in the production of transgenic organisms.

In this chapter, you will explore how biotechnology, including specific genetic techniques, is used and how it can affect **biodiversity**. It is also important to assess the benefits and risks to make valid social and ethical decisions about using biotechnology, in order to limit its impact on living things and ensure that the benefits outweigh the harm.

8.1 Biotechnology – past, present and future

Biotechnology has been in use for many thousands of years, despite the fact that it was only named relatively recently. We call the practices from thousands of years ago *ancient biotechnology*. Those in use from the late 1800s to the mid-1900s are called *classical biotechnology*, and those implemented since the

discovery of DNA (1950s to the present) are known as *modern biotechnology*. The term 'biotechnology' was introduced in 1919 by a Hungarian scientist, Karl Ereky, to refer in general to processes where raw materials were converted into useful products, such as on industrial farms.

Most biotechnological developments in early times were based on using products available in nature to improve living conditions and survival, targeting the basic human need for food, shelter and clothing. Today's biotechnology has similar improvements in mind, but the techniques are implemented at a more precise molecular level, some with far-reaching consequences.

Past biotechnology

Worksheet
Ancient biotechnologies

Ancient biotechnology was in practice for thousands of years without a full understanding of the biological and biochemical processes involved. Practices included the use of yeast to make bread, as well as beer and wine fermentation. Foods such as yoghurt and cheese were produced using bacterial cultures.

Classical biotechnology emerged with the contributions of scientists such as Louis Pasteur and Gregor Mendel. Pasteur discovered that fermentation was not a chemical process but a biological one. Mendel introduced the idea of genetic information being transferred from one generation to the next, a concept central to biotechnology today. By the end of the 19th century, cell products such as enzymes were in use. The first antibiotic was discovered in 1928 and antibiotics were used for the first time as medicine in 1941. Biotechnology was moving from the farmyard and the garden into the laboratory.

Present biotechnology

Modern biotechnology took a huge step forward with the discovery of DNA and its subsequent manipulation at a molecular level. This has been likened to the biological equivalent of humans landing on the moon. Manipulation of DNA to create products for human use is known as **gene technology**. This is a branch of biotechnology where the end products are very precisely obtained, because people now understand how DNA makes proteins, such as enzymes, which control all aspects of cell metabolism. Modern biotechnology uses the tools of **genetic engineering** not only to make specific proteins, but also to regulate cell processes so that specific outcomes can be achieved and precise needs can be met.

The applications of biotechnology have varied over time, with specific uses in agriculture, medicine and environmental remediation. Some scientists have tried to classify the various branches of biotechnology using colours – *green biotechnology* focuses on developments in agriculture, *red biotechnology* on medical and pharmaceutical applications, *white biotechnology* on industrial purposes, and *yellow biotechnology* on food production. However, the branches tend to overlap – for example, genetically modified crops are developed in agriculture but are used to increase food production – and so the classification system still requires some refining.

Traditional applications of biotechnology were mainly in plant and animal breeding. The growing of crops was the beginning of agriculture, with humans attempting to create a stable supply of nutritious food for their clans. The domestication of animals provided humans with help in doing physical work, and had the added benefit of protection. As far back as 15000 BCE, as seen in evidence of the oldest dog fossils from Siberia, humans were using dogs for these purposes.

FIGURE 8.2 *Canis lupis*, the Eurasian grey wolf, is believed to be the ancestor of the modern dog, domesticated 15 000–16 500 years ago.

Modern applications of biotechnology still encompass food production and industry, for example the production of genetically modified (GM) food and the production of industrial products, such as paper. While animal breeding still plays a large role, there has been a shift in breeding techniques from selective breeding to genetic modification of animals. A further broad application of modern biotechnology is in medicine and pharmaceuticals to improve human health. Environmental conservation is another global focus of biotechnology today.

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Future biotechnology

In future applications, advances in genomics as well as our understanding of how proteins are expressed in cells will be used for improving the treatment of infectious diseases, cancer and other genetic disorders. *Green chemistry*, where proteins can be modified to produce environmentally friendly products, is being explored. Cellular metabolic pathways are being manipulated to enable a range of outcomes, including the production of edible vaccines using micro-organisms, the manufacture of synthetic cells and organs, and the transplantation of organs from animals into humans.

Ancient biotechnology

Early humans obtained their food by hunting and gathering. Later, they began raising their own animals and growing their own crops.

Agriculture

Farming began in the Middle East, known as the 'fertile crescent', approximately 10 000 years ago. By 7000 years ago, farming was established in China, and by 5000 years ago in Mesoamerica (central America and the southern tip of North America). Over hundreds of years, humans worked to improve the quality and yield of their food, realising the advantage of selecting seeds from the best crops and breeding the best-quality animals. This was the start of **selective breeding** or *artificial selection*, involving the human 'manipulation' of living organisms. People selected organisms that they wished to cross-breed, ensuring that selected individuals possessed desirable characteristics that could be passed on to future generations. Cross-breeding different varieties of organisms resulted in stronger, healthier offspring than inbreeding – a phenomenon known today as **hybrid vigour**.

The shift away from hunting–gathering towards farming resulted in food surpluses, which meant an increase in population density and, for the first time, a significant change in biodiversity.

Aboriginal people and aquaculture

Archaeologists have found stones and foundations, up to 6000 years old, that are evidence of a canal system built by Aboriginal people. The traces show that canals meandered from the ocean to inland areas in the Lake Condah district of western Victoria. There is also evidence of eel traps (known as Budj Bim eel traps) being used in the district. There is similar evidence in Mount William and Tolondo, where canals were built to connect naturally occurring swamps, and in the Barwon–Darling river system in western NSW. Aquaculture has continued among the Aboriginal people (Fig. 8.3), and elders still recount stories of their grandparents' days about eel traps – how they were used, and

Aboriginal and Torres Strait Islander histories and cultures

(see State Library of NSW (IE1676894), Photo by Thomas Dick

FIGURE 8.3 Aboriginal man collecting oysters, an example of early aquaculture

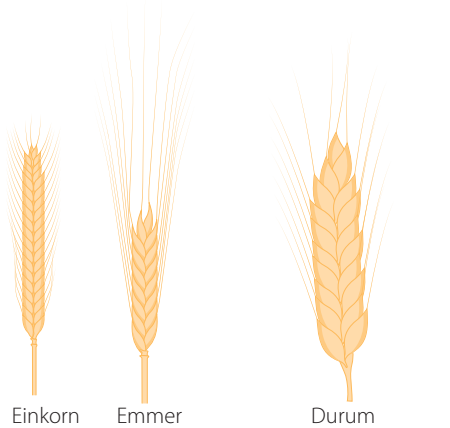

FIGURE 8.4 Comparison of ancient wheats and modern wheat

the preservation of eels by smoking them in the hollows of trees, to sustain people when food was scarce and for trade with other groups.

Food production: bread and cheese making

After domestication of food crops and wild animals, humans began to implement other biotechnologies, such as cheese making and bread making. Some of the earliest evidence of changes in plants due to domestic breeding comes from ancient evidence of the use of wild grains in caves in the Middle East, inhabited approximately 10 000 years ago. The layers of fossilised grain reflect a change in the characteristics of the grain, with the older wild grains of Einkorn wheat in the bottom layers having to be pounded to release the seeds, whereas the younger Emmer wheat grain in the top layers could naturally release the seeds (Fig. 8.4).

Bread making

Images carved in the walls of tombs in ancient Egypt depict the process of bread making (Fig. 8.5). This explains the need for wheat from which seeds could be naturally extracted. Evidence of the use of yeast to make bread is seen in Egyptian tombs from around 2000 BC. It is assumed that wild yeast made its way into dough by accident around 10 000 years ago and was later intentionally introduced when the benefits of its actions were noted. Yeast ferments sugars to produce carbon dioxide, which in turn makes the bread rise. Yeast was extracted from malt-wort and added to the dough. Baker's yeast has been used since the 1920s and today the *Saccharomyces cerevisiae* strain is used to ferment bread dough.

Caves containing fossilised wheat were also rich in animal remains, indicating a change in the types of animals eaten. As humans moved from being a migratory species to settled groups, they learned how to use the plants and animals around them. Maize, beans and potatoes in Central America, rice, millet and buffalo in Asia, and wheat, sheep and goats in the Middle East were bred and farmed.


Heritage Image Partnership Ltd / Alamy Stock Photo

FIGURE 8.5 Bread making in ancient Egypt

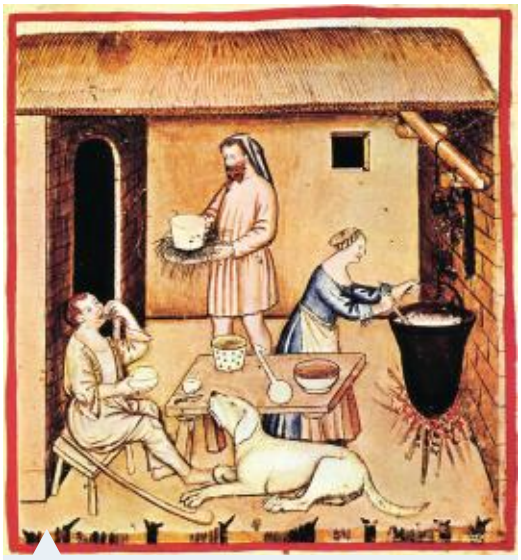
ART Collection / Alamy Stock Photo

FIGURE 8.6 A meal with cheese, from an 11th century medieval handbook on health and wellbeing

Cheese making

Cheese making involves the action of bacteria on milk and dates back as far as 10 000 years ago. The process was improved over the years, and by the time classical cheese making was in progress, people understood that enzymes could be purposely added, and when the enzymes combined with bacteria naturally present in the air, curds and whey were produced. Pressing of the curds removes

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excess water, and storage in a dry environment allows ripening, which often includes the action of other surface bacteria that give the cheese a distinct flavour.

Evidence of yoghurt making dates back to approximately 4000 BC. The Chinese used *Lactobacillus* to ferment milk into a semi-liquid consistency, extending the time over which the milk could be stored and used. This same genus is still used to make yoghurt today.

Classical biotechnology (1800s to mid-20th century)

At particular points in time, key findings have led to remarkable advances in the field of biotechnology. One example of this is the discovery by Louis Pasteur in 1857 that fermentation results from the action of micro-organisms.

Fermentation

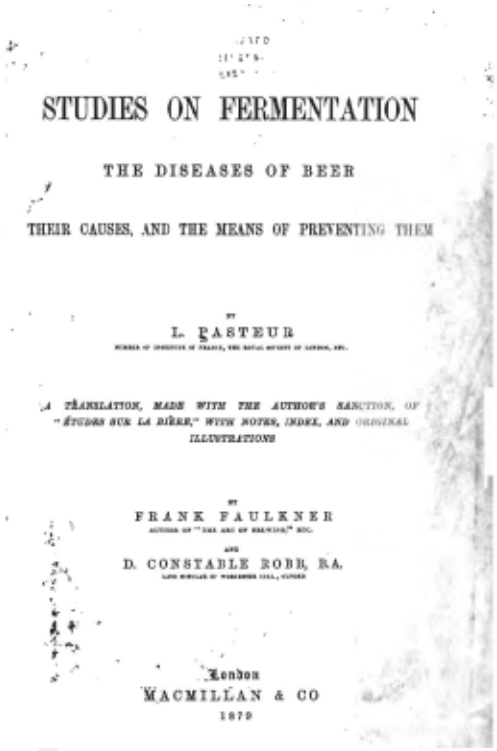
The word **fermentation** is derived from the Latin word *fervere* – 'to boil'. The use of fermentation began over 6000 years ago. It is believed to have started in ancient Egypt, when bread or milled grain left in a damp place began to ferment due to yeasts present in the air. The wheat would have provided sugars produced from starch, and the yeast from spores in the environment would have used the sugars to ferment the bread and produce alcohol.

The discovery that the process of fermentation resulted from use of the biological organism yeast is attributed to the work of Joseph Louis Gay-Lussac and Louis Pasteur. Although people had been using these processes for thousands of years, it was not until the 1600s that it was recognised that the froth on beer was due to gas accumulation. Gay-Lussac proposed the chemistry behind this, but the discovery that microbes were responsible for the gas production is attributed to Pasteur (Fig. 8.7). In the 1860s, Pasteur used experimentation to show that the fermentation process is biological, not chemical.

Reread Chapter 3 of *Biology in Focus Year 11* on alcoholic and lactic acid fermentation (anaerobic respiration).

Classical biotechnology therefore uses known biological material such as cells (for example, yeast and other microbes), as well as known cell products (such as enzymes and antibiotics) to achieve a particular goal.

Brewers used barley and other grains to make beer. Hops were added to the process in the Middle Ages, and the process relied on yeast from the air. A sugar-rich solution that contains either grain or fruit naturally contains yeasts and, if allowed to stand, the yeast ferments the sugars to produce alcohol and carbon dioxide. In wine making, the skin of grapes contains natural sugars and yeasts that facilitate the process of fermentation.


STUDIES ON FERMENTATION

THE DISEASES OF BEER

THEIR CAUSES, AND THE MEANS OF PREVENTING THEM

BY

L. PASTEUR

A TRANSLATION, MADE WITH THE AUTHOR'S SANCTION, OF "ÉTUDES SUR LA BIÈRE," WITH NOTES, INDEX, AND ORIGINAL ILLUSTRATIONS

BY

FRANK FAULKNER

AND

D. CONSTABLE ROBB, B.A.

London

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1879

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FIGURE 8.7 Pasteur's findings on fermentation were published in 1879.

Medicine and antibiotic production

The use of plants to heal the sick predates modern medicine. Many conventional drugs and medicines used today are derivatives of ancient remedies. Willow bark, which contains salicin, was used to treat inflammation and for pain relief. Turmeric was used to improve circulation, and its anti-inflammatory properties are recognised today. The Persians and Egyptians eased pain by using the milk of the opium poppy (Fig. 8.8). Poppy seed cakes and poppy pods have been found in Neolithic Swiss dwellings from 4000 years ago. Bark from the *Cinchona* plant, also known as Jesuit bark, was used by indigenous people in South America to ease fever. The plant contains quinine, which has antimalarial properties.

A growing awareness of medicinal plants from ancient times to today has led to contemporary scientists creating medicine from plants and fungi, used in modern pharmacological drug development. One of the most famous natural products discovered was an antibacterial

FIGURE 8.8 The milky sap of *Papaver somniferum*, the opium poppy, is used to make opium, and its seeds are used for culinary purposes.

product made by the fungus *Penicillium*. The product was discovered in 1928 by English bacteriologist Alexander Fleming. It was later stabilised by Australian pharmacologist and pathologist, Howard Florey, to form the antibiotic known today as penicillin. This application revolutionised the treatment of infectious diseases caused by bacteria, and penicillin is still used today.

Plant selective breeding techniques

Hybridisation and artificial pollination were, and still are, commonly used in horticulture and agriculture to improve the quality of plants being grown or raised. Hybridisation produces hybrid offspring with combined traits that make them better suited to their environment or for their end use (such as bigger flowers, higher nutrient value, increased crop yield, shorter breeding time, greater resistance to drought, disease or salinity, or higher stamina), termed 'hybrid vigour'.

At times, traits that are beneficial to breeders and consumers may be detrimental to the plant. For example, making fruit sweeter may attract more feeding by insects and other pests. Some traits that increase a plant's resistance to disease may be lost when the plant is bred to enhance other traits.

KEY CONCEPTS

- Biotechnology is the use of living organisms or their products for human benefit.
- Ancient biotechnology began thousands of years ago, with the domestication of crops and animals and the use of living cells to make food products such as bread, cheese and wine.
- Classical biotechnology came about with advances in scientific understanding of the role of living organisms in fermentation (Pasteur), antibiotics (Fleming) and genetics (Mendel).

CHECK YOUR UNDERSTANDING 8.1a

1. Define 'biotechnology'.
2. Identify the biological materials that may be used in biotechnology.
3. Name three fields of biotechnology, classified according to 'colour'.

Modern biotechnology (1950s – present)

Worksheet
Technologies to manipulate DNA

Modern biotechnology to a large extent involves **genetic engineering** – techniques that are used to manipulate DNA to meet the needs of society, the environment and science in a very precise way.

Genetic engineering involves humans manipulating the pattern of bases in the DNA (genotype) of an organism to change its phenotype, making the individual appear or act differently. The most common steps in genetic engineering involve 'cutting, copying and pasting' DNA sequences, a different version of what you may do on a computer to a word processing document. An example of genetic engineering is where scientists have modified bacteria by using enzymes to snip out (cut) a human gene and insert (paste) it into bacteria, enabling the bacteria to produce human proteins such as insulin and human growth hormone. The new DNA, made up of DNA from more than one species (bacterial DNA with a human gene inserted), is known as **recombinant DNA** (rDNA) (Fig. 8.9). These organisms are called **genetically modified organisms (GMOs)** or, if they are able to pass on their newly constructed genome to the next generation, **transgenic species**.

You will learn more about transgenic species in Chapter 9.

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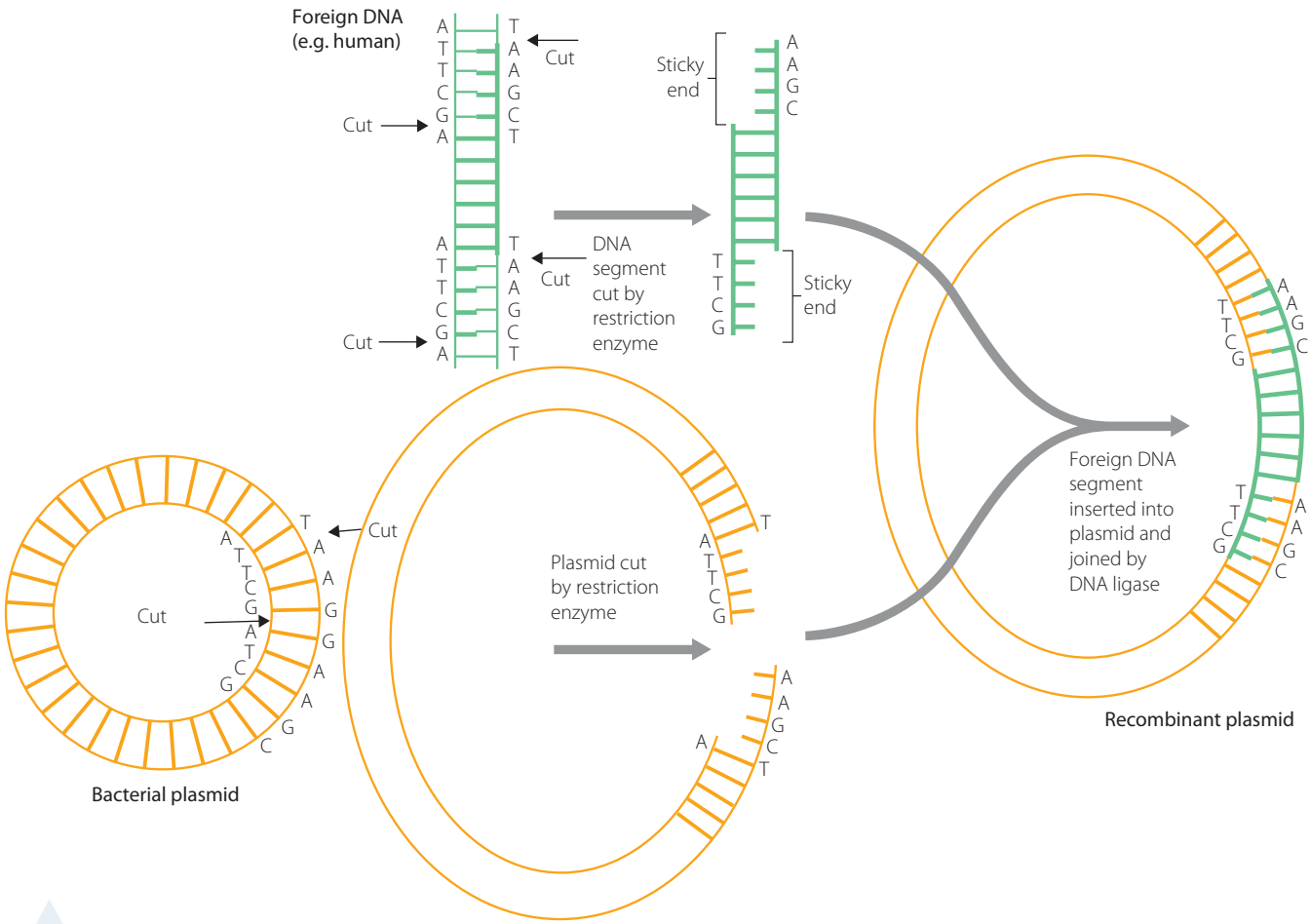

FIGURE 8.9 Formation of recombinant DNA in the bacterium *E. coli*

DNA technology is a generic term for the use of specialised biological tools to modify, measure, manipulate and manufacture DNA. All these 'tools' (many of which are enzymes) come from other living organisms, such as bacteria and viruses.

Some of these genetic engineering techniques are dealt with in other chapters, so only a brief outline is provided below, to clarify how they are used sequentially and applied to achieve specific outcomes in modern biotechnology.

Technology to manipulate DNA

DNA splicing

DNA splicing means cutting out genes. The required gene or sequence of bases in a DNA molecule is spliced out, using restriction enzymes that cut DNA at specific base sequences (Fig. 8.9). Restriction enzymes occur naturally in bacteria. Their purpose in nature is to 'chop up' foreign DNA from invading viruses. Scientists use them like 'molecular scissors', snipping DNA into smaller pieces at specific base sequences (Fig. 8.10).

See Chapter 9 for more detail on this technique.

FIGURE 8.10 **a** Sticky ends produced by cutting DNA with the restriction enzyme ECoR1; **b** blunt ends produced by cutting DNA with the restriction enzyme Alu1

DNA amplification is dealt with in Chapter 9.

DNA amplification

DNA amplification means copying genes. This is done through a process known as the polymerase chain reaction (PCR), in which the DNA polymerase enzyme is used to replicate DNA fragments many times before they are inserted into the new genome.

Recombining DNA

Recombining DNA means 'pasting' genes. A DNA ligase enzyme is used to join pieces of DNA together, forming bonds in the sugar-phosphate backbone of DNA.

Technology to analyse and visualise DNA

DNA molecules are too small to be seen, even with a microscope, so special techniques must be used to enable scientists to analyse and visualise them. DNA is usually cut into small fragments that can then be identified and built into a picture of a whole DNA molecule or genome. DNA analysis and visualisation techniques include gel electrophoresis and gene probes.

Agarose gel electrophoresis

Gel electrophoresis (Fig. 8.11) is used to analyse DNA and then identify the 'DNA fingerprint' of an individual (described in Chapter 6, page 197). DNA is fragmented and passed through a gel. The distribution pattern of the fragments can be seen as bands on the gel, each band representing a DNA fragment of a particular size (Fig. 6.6). This information can be used in a manner similar to a bar code, to identify an individual or a species.

FIGURE 8.11 A researcher injecting genetic material into an agarose gel

Gene probes

A gene probe is a specific length of single-stranded DNA (20–1000 nucleotides in length) that is complementary to a known DNA sequence from a specific gene. Gene probes can be manufactured artificially and tagged with a fluorescent dye or radioactive atom so that the probe and the DNA that it binds to can be seen. This allows the DNA to be 'visualised'. Thousands of genes can be tested at a time, using a micro-array.

DNA sequencing

DNA sequencing is used to determine the exact nucleotide sequence of DNA or a gene, to find (visualise) the genetic code for a particular phenotype. This may be done using gel electrophoresis or using 'next-generation' automated

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sequencing technologies, such as nanopores (Chapter 6). All sequence data that is published is saved in a database called GenBank for use by the public.

DNA profiling

DNA profiling involves DNA amplification of short tandem repeats (STRs) by PCR, followed by gel electrophoresis. Its purpose is to compare the base sequences of two or more individuals to determine relatedness based on differences in the length of DNA repeats.

FIGURE 8.12 In DNA profiling, DNA sequences are analysed

Applications of modern biotechnology and social implications

Human needs drive biotechnology, which today still includes the creation of food, medical and agricultural products, and has also extended to applications in industrial biotechnology and gene technology, resulting in new varieties of living organisms. This has impacts on Earth's biodiversity.

Worksheet
Modern technologies

Industrial biotechnology applications

Industrial biotechnology includes fields such as enzyme engineering, bio-nanotechnology and synthetic biology, as well as biochemical and bio-material engineering.

Applications of industrial biotechnology include:

- *pollution prevention*, for example the use of microorganisms to clean up and reduce waste; the use of enzymes in washing powders to remove stains, replacing phosphates that were previously used and built up in water systems, contributing to eutrophication (overgrowth of plant life in waterways); the production of environmentally friendly products such as fertilisers, biopesticides, biofuels and biodegradable materials made from plants
- *biomaterial production*, which involves any natural or synthetic substance being made to interact with biological systems for a medical, therapeutic or diagnostic purpose. Examples include joint replacements, artificial heart valves, stents and breast implants.

Future applications include:

- *biofabrication* – the automated production of tissues and organs, using the principles of 3D printing and materials such as cells, fibres and gels, to replace diseased or injured tissue
- *synthetic biology* – using computer technology to construct synthetic genomes that can function in a living cell. In 2010, American scientists J. Craig Venter and his team created the first synthetic cell – a microbe. They used a computer to create a digital genetic code of 1.1 million base pairs. The genome, a replica of the full genome of the bacterium *Mycosplasma mycoides*, was made in the laboratory from synthetic nucleotides. When implanted into an enucleated cell of another related bacterium, the genome directed the bacterium to produce new proteins. The synthetic cell divided under the instruction of the synthetic code to become a colony of *M. mycoides* cells. The cell was nicknamed 'Synthia' and is acknowledged as the first synthetic cell. This new biotechnology opens up a world of possibilities. The application of making large-scale changes to codons in genetic systems using computers will enable different and/or novel amino acids to be incorporated into proteins, to improve gene expression and exclude genetic diseases.

See Chapter 9 for more information on applications of biotechnology in industry, resource conservation and agriculture.

Weblink
Industrial biotechnology
Analyse the table of products and summarise the benefits to society and the environment of at least three of these products.

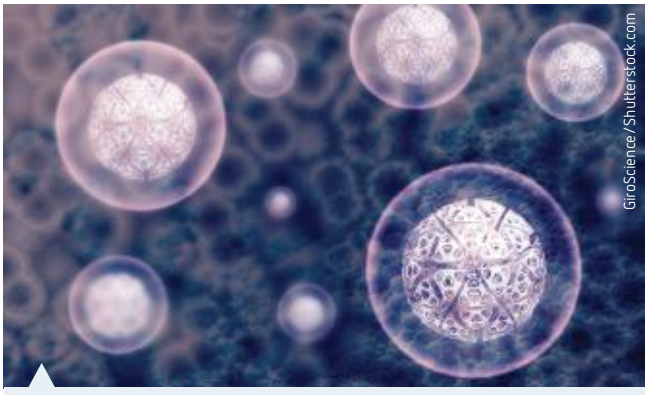

FIGURE 8.13 Synthetic cells are built entirely through genetic engineering.

Agricultural biotechnology applications

Applications of agricultural biotechnology include improving plant and animal production by increasing yield, nutritional value and resistance to disease. Agricultural biotechnology aims to enhance the quality of, and economic returns from, commercial crops by using reproductive technology and gene technology (Chapter 9).

Sustainability

Ethical understanding

Asia and Australia's engagement with Asia

Weblink
The latest defence against animal extinction

Reproductive technologies and genetic diversity

Modern-day reproductive technologies include artificial insemination (animals) and pollination (plants), in vitro fertilisation, embryo transfer and cloning. Some of these advanced technologies use DNA manipulation techniques to insert desired genes into organisms by genetic engineering, creating transgenic species (Chapter 9).

Advantages of selective breeding in agriculture currently include increased:

- milk yield in female cattle
- quality of beef in cattle
- size of eggs from chickens (and more frequent laying)
- grain yield from wheat
- protein in food
- vitamin content in food (for example, golden rice).

marekuliasz/Shutterstock.com; Marius C/Shutterstock.com; A G Baxter/Shutterstock.com

FIGURE 8.14 Biodiversity is one of the concerns of agricultural biotechnology.

Agricultural applications of biotechnology include the use of genetically engineered crops, such as golden rice, to address hunger in poor and developing countries and work towards eradicating starvation. Golden rice was created by the insertion of two genes, one from the daffodil (replaced by corn in a later version) and one from a bacterium, into a rice genome. Golden rice differs from other rice in that it contains a vitamin A pre-cursor, beta carotene. Once this rice, with its much higher nutritional (vitamin A) content, was developed, its creators formed a deal with a company to produce it in large quantities for humanitarian purposes, to help to combat vitamin A deficiency (which can cause blindness and other serious health problems) in the developing world.

However, worldwide concern has grown about loss of genetic diversity and biological resources, due to farming generally and, in particular, farming in developing countries. At a convention on biological diversity in Japan in 2010, a new protocol (the Nagoya Protocol) was adopted. This addressed a further issue of access to genetic resources by various countries and the 'fair and equitable sharing of benefits' from the use of these resources. As a result, a legal framework was set up for the biotechnology industry to implement a common goal of sustaining biological diversity and for benefit-sharing obligations in the Convention on Biological Diversity (CBD).

Conservation biology applications

Progress has also been made in using plant biotechnology for biodiversity conservation. Advances in **genomics** and **proteomics** have enabled researchers to identify desirable traits in crops and plants using molecular markers, and to bank (save) favourable germplasm. **Germplasm** is any living tissue from which new plants can form, and includes whole plant plant parts (such as leaves or stems), pollen and clusters of cells. Germplasm can be incorporated into crop varieties, either through selective breeding or using genetic manipulation techniques, to increase biodiversity.

Applications of animal biotechnology include researchers mapping relatedness between animals (in the wild and in captivity) and then designing breeding programs to maximise hybrid vigour and genetic biodiversity (Fig. 8.14).

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Artificial insemination is being used for animal conservation in the wild and in zoos, to maintain biodiversity. For example, researchers at the Smithsonian National Zoo in the USA have used artificial insemination in animals at risk of extinction, producing one hundred black-footed ferrets that could then be released into their natural former habitat.

Veterinarians at the Zoological Society of London's conservation centre have been conducting habitat surveys in Niger and Chad in Africa to try to reintroduce a herd of antelope (scimitar-horned oryx) that are extinct in the wild. The zoo has appealed to other zoos around the world to provide either female oryxes or oryx sperm so that the resulting offspring have the broadest possible genetic variation.

FIGURE 8.15 The Sumatran rhino, almost extinct in the wild, is part of a captive breeding program.

Some species that are close to extinction, such as the Sumatran rhino (Fig. 8.15), cannot be saved in the wild. Captive breeding technologies are critical in saving these species.

The southern white rhino has been brought back from the brink of extinction by captive-breeding programs. At the start of the 20th century, there were approximately 50 southern white rhino left. As a result of captive breeding with close mapping of relatedness, there are now over 20 400 white rhinos, with no apparent birth defects despite inbreeding from a small parent population. The rhinos remain in captive populations in conservation centres, to reduce the risk of them being eliminated in one place by disease, natural disaster or poaching.

Banks for animal genetic material have also been established, and oocytes and gametes are being cryogenically preserved at zoos around the world, to conserve animal genes for biodiversity purposes.

Genetic engineering can also produce plants and animals that are more fertile and resistant to disease and drought, and plants that can grow in nutrient-poor soils. This is crucial in maintaining biodiversity.

Weblink
Saving the rhino

Weblink
Biotechnology for biodiversity
Read about DNA banks for plants and the use of molecular markers.

Medical technology applications

Gene therapy is a technique based on delivering normal genes that function correctly, to individuals who are lacking a functional copy and as a result suffer from a genetic disorder. For example, scientists have had success with the disorder called severe combined immunodeficiency (SCID). Sufferers are very susceptible to infection, as the mutant gene disrupts the functioning of the B and T cells in their immune systems.

See Chapter 9 for more information on gene therapy and Chapter 12 for more information on B and T cells.

In vitro fertilisation has been in use in humans for about 40 years. Over this time it has become more sophisticated and now has a greater success rate, thanks to improvements in scientists' understanding of endocrinology and reproduction in various species, and advances in biotechnology. It is not only successful in allowing couples who have fertility issues to reproduce, but it is also being used to prevent children being born with inherited genetic diseases – one technique uses gamete material from three parents to avoid a child being born with mitochondrial disease. (See the weblink.)

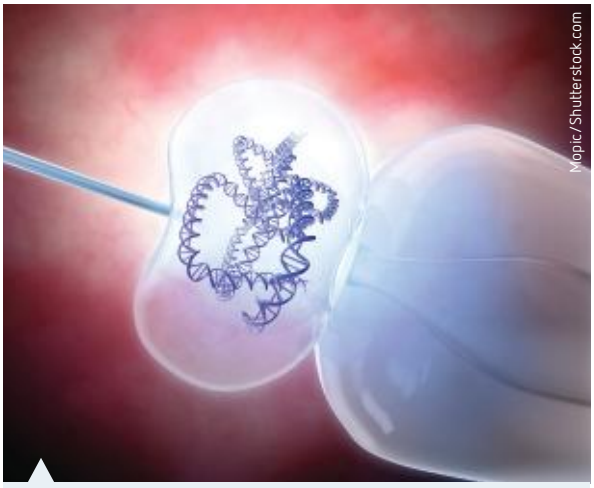

FIGURE 8.16 Gene therapy involves the manipulation of DNA to overcome genetic disorders.

Weblink
Three-parent technique
Read about babies conceived using DNA from three people, to prevent mitochondrial disease.

INVESTIGATION 8.1

Secondary-source investigation into the uses of biotechnology: past, present and future

Critical and creative thinking

Information and communication technology capability

In the past, people developed biotechnologies to improve the availability and quality of food, from growing crops to making products such as bread, cheese, beer and wine. Traditional biotechnology progressed as scientific understanding of these processes improved and knowledge and understanding of cell and molecular biology increased. Biotechnology has made major advances, enabling scientists to manipulate genes and their products more precisely than ever before and applications have moved into the realm of gene therapy in medicine.

AIMS

To gather data about the development of biotechnologies from past to present
To analyse trends in biotechnology developments and identify the scientific knowledge and understanding needed for each breakthrough

METHOD

1. Working in groups of three, you are to create timelines of biotechnological developments in particular fields. You will then compare your different timelines within the group, to identify trends and patterns common in major breakthroughs.
2. Individual work: select two applications of biotechnology (examples below) and conduct research, gathering relevant information from a variety of sources, to follow the development of biotechnologies from ancient times to the present. You must select at least one biotechnology that is not being researched by another member of your group. Extrapolate at least one future direction that research in this field is taking.
3. Create a timeline to record your information, placing the two biotechnologies on each side of the line for comparison.

Some examples of applications of biotechnology that you may wish to research are those used for:

- food production – from ancient agriculture/aquaculture to current genetically modified food
- fermentation – from ancient practices to applications of modern fermentation
- selective breeding – from domestication to genetically engineered organisms
- medical /pharmaceutical applications – from the ancient practice of using bark to gene therapy today
- environmental applications – from early agriculture to harnessing nitrogen, phosphorous and biofuels today
- biodiversity conservation – from agricultural beginnings to plant DNA banks to reintroducing genes of extinct animals into the gene pool to increase biodiversity.

RESULTS

Record your results in the form of a timeline, including dates (or a range of dates where applicable) and events in the development of the applications of biotechnology that you are researching, arranged in chronological order. The timeline may be drawn from the bottom to the top of the page (earliest date at the bottom), or from left to right on a landscape-oriented page (earliest dates on the left).

DISCUSSION

As a group, analyse your timelines to find common trends in the development of biotechnologies throughout history and identify the scientific knowledge and understanding needed for each breakthrough.

CONCLUSION

Based on your research, sum up the advances in biotechnology in your selected field and outline the scientific knowledge and understanding required for breakthroughs in these fields.

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KEY CONCEPTS

- Modern biotechnologies have been developed to manipulate, analyse and visualise DNA in genomes.
- Applications of modern biotechnologies include:
  - industrial uses: the production of biopesticides, the use of enzymes in pollution reduction, using cell products to make fertilisers
  - agricultural uses: reproductive technologies for selective breeding and genetic modification of foods
  - medical uses: gene therapy and human IVF.

CHECK YOUR UNDERSTANDING 8.1b

1 Distinguish between ancient, classical and modern biotechnologies, giving an example of each.
2 Create a table to outline two uses and applications of biotechnology, one past and one present, in each of the following areas:
   a agriculture **b** food production **c** medicine or pharmaceuticals.

8.2 Ethics and social implications

Ethical understanding
Civics and citizenship
Sustainability

The manipulation of DNA by humans is a contentious issue, as it raises ethical questions – people need to make wise decisions when important values are at stake. Selective breeding may benefit society by making agricultural practices more productive, but when the principles of selective breeding are applied to humans there is the potential for breaches of ethics – attempts to create a race of humans with 'superior' genetic qualities, for example, as was seen during World War II. In the late 20th century, some drug trials breached the principles of research ethics by using volunteers who were vulnerable or terminally ill, subjecting them to harm. DNA manipulation techniques in use today have potential for misuse in biological warfare and terrorism. New biotechnologies are powerful and have great potential to change the genomes of organisms, with global impact.

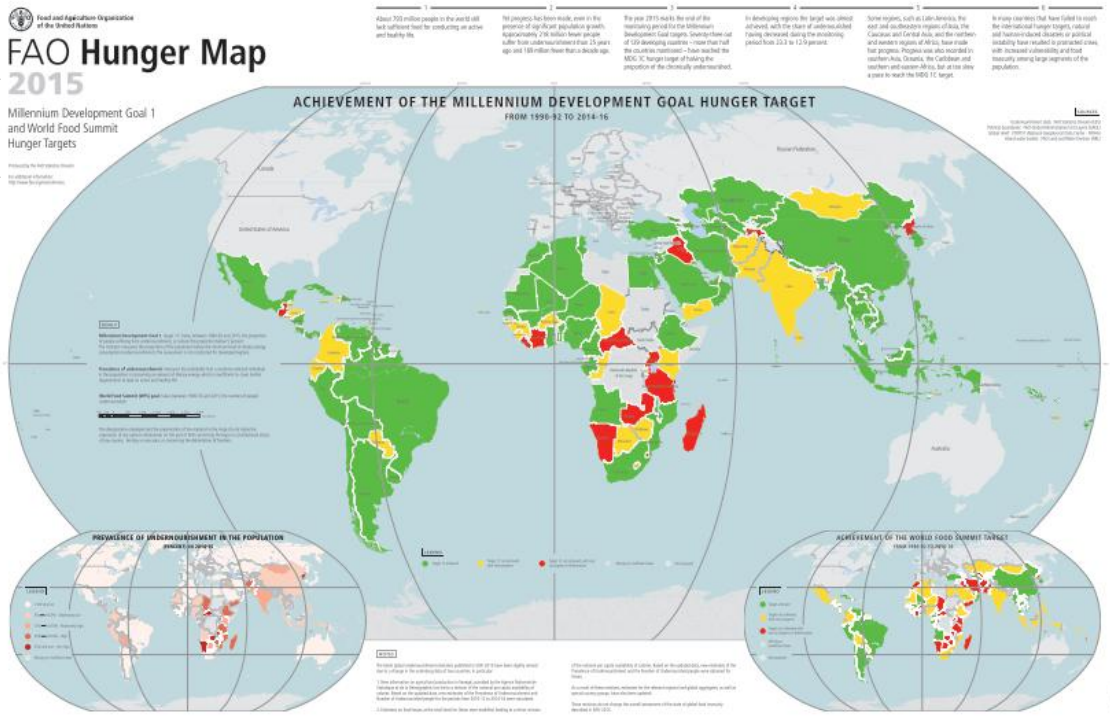
Technologies such as cloning and the CRISPR technique (page 275) allow very precise introduction of genes into cells, making it easier for scientists to alter the human genome and combine the genes of different species. This raises issues of human and animal rights and freedoms, including the question of how 'tampering with nature' may alter the path of evolution. We need to consider our responsibilities carefully, to ensure that these technologies maximise benefits, preserve privacy, and support dignity and informed consent, while minimising harm. For example, positive inroads are being made in using biotechnology in conservation, increasing the biodiversity of animals in farming and in the wild and to further research, but what are the costs to society and the environment?.

Case study: Global need for increase in agricultural products

The United Nations, Department of Economic and Social Affairs, Population Division (UNESCO) has estimated that, by the year 2050, there will be an additional 2.41 billion people in the world to feed (UN-DESA-PD 2011). The demand for food and other resources to sustain the population will grow exponentially and this puts new demands on the agricultural industry (Fig. 8.17).

This demand for food is not spread evenly across all nations. It is predicted that there will be very little increase in human populations in the more developed regions of the world (Europe, Northern America, Australia/New Zealand and Japan), but very high increases in population are likely in less developed countries (Asia, South-East Asia, Africa).

Reproductive and genetic technologies in livestock and crop production can make a positive contribution in developing countries, alleviating poverty and hunger, reducing the threat of disease and ensuring environmental sustainability.



Source: Food and Agriculture Organization of the United Nations, 'FOA Hunger Map 2015', Millennium Development Goal 1 and World Food Summit Hunger Targets. (http://www.fao.org/publications/card/en/c/1a001c07-6567-4c0a-b5ca-b5b86bc0e881/). Reproduced with permission.

FIGURE 8.17 United Nations world food hunger map. Green areas in the large map indicate that Millennium Development Goal Hunger Target 1C (to halve the proportion of people suffering from undernourishment or reduce this proportion to below 5 per cent) has been achieved; yellow areas indicate target not achieved and slow progress; red areas indicate target not achieved and no progress or worsening.

Part of the solution to food shortages is to improve agricultural productivity by increasing the:

- amount of food – larger numbers of plants and animals farmed for consumption
- quality of food – greater amount of edible protein in foods (crops and livestock).
- resistance of some crops and livestock to disease, drought and floods.

As knowledge and technological power in the biological sciences grow, the need arises for careful consideration of the values that are at stake. An International Bioethics Committee has been formed by UNESCO to ensure that progress in genetics is accompanied by reflection on ethical and legal issues. They also aim to take action to heighten awareness of human dignity and freedom of choice and to ensure respect for all living organisms and the environment. They encourage countries to work together to reach international agreement on legal and ethical issues in molecular biology.

Ethical understanding

Critical and creative thinking

When analysing the social and ethical implications of new biotechnologies, the following should be considered:

- medical and health benefits
- financial and social justice issues
- animal and human rights
- effects on the environment.

Personal and social capability

Making an ethical decision

Weblink
Regulation of gene technology in Australia

Bioethics refers to the study and investigation of how decisions in medicine and science affect society and the environment. We need to take into account the benefit or harm to the lives and health of individuals and society as a whole. We also need to consider justice and equity – fair and equitable treatment for all. When faced with a controversial issue, people can differ in their opinion of what is right or wrong, which may lead to a moral dilemma, where trying to decide what is right or wrong challenges a person's basic beliefs.

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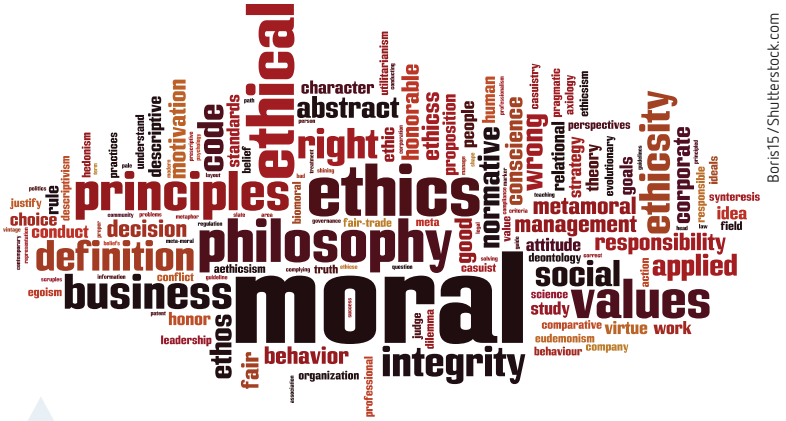
John Warburton-Lee Photography/ Alamy Stock Photo

FIGURE 8.18 Women carrying farming produce in a developing country



Boris15/Shutterstock.com

FIGURE 8.19 Aspects of bioethics

TABLE 8.1 Basic principles of ethics

	PRINCIPLE	QUESTIONS	AIM
1	Benefit and non-harm	In whose best interests is the action – does it benefit individuals/groups/society in general/future generations? How reliable and useful is the procedure? Do the advantages outweigh the risks?	Wellbeing
2	Individual rights and autonomy	Is the person properly informed of the advantages and harms or risks? Does the person have freedom of choice? Are the rights of others protected or breached?	Choice
3	Privacy and societal perception	Will the person's genetic information remain private? What is the right thing to do if a breach of privacy could save another person's life? How might the release of this information affect the way in which society or another individual perceives that person?	Respect
4	Equity and justice	Is there equal access for all members of society (no bias in terms of gender, socioeconomic status or ethnicity)? Is there fair sharing of resources that can be justified? How can discrimination be avoided?	Fairness

To become informed, reflective and responsible citizens, we need to develop our skills in critical thinking. The media, politicians and religious institutions often give wide exposure to controversial issues, and we do not always want them to make decisions on our behalf. Ethics are the principles that help us decide what is the right thing to do.

Scientists use *evidence* and *reasoning*, rather than emotion, to understand and respond to technological advancement and scientific developments. We need to weigh up the risks and benefits of using certain technologies or procedures, and compare these with the potential consequences of not using the procedures. This process of reasoning should help us gain an informed ethical standpoint that we can justify. However, we also need to take into account our own beliefs, values and cultural experiences. As a result, bioethicists often use frameworks that have been created to guide thinking and decision making. The frameworks that are selected for use may differ, but they serve a common purpose – to assist the users to evaluate conflicting points of view and reach a judgement based on logic and reasoning.

Today many people develop careers as bioethicists. They work towards developing guidelines to assist individuals, healthcare providers, conservationists and global communities to make decisions about biological issues that pose moral dilemmas. Bioethicists come from a variety of backgrounds, including lawyers, scientists, doctors, nurses, philosophers, social workers, theologians and educators. They may be employed by governments, hospitals and universities. Although bioethicists may disagree on certain points of view among themselves, they generally agree on a basic set of guidelines, referred to as *the basic principles of ethics*. These principles are summarised in Table 8.1, along with questions to help users decide whether a procedure is ethical.

INVESTIGATION 8.2

Secondary-source investigation to research an ethical issue

Critical and creative thinking

Information and communication technology capability

Sustainability

Ethical understanding

Civics and citizenship

Personal and social capability

BACKGROUND INFORMATION

Many people regard golden rice as an example of how biotechnology can be used to help developing nations, while others think it is a risk to humans and the environment. Questions of whether resources such as golden rice should be privatised by companies with intellectual property rights have further fuelled the debate. Are there other ways to address this issue that would promote the use of genetic engineering to alleviate hunger in developing nations? Traditional technologies worked for some nations – for example, the large-scale use of fertilisers and pesticides, as well as improved seeds and livestock, increased agricultural production in the USA and Europe to a level where there is more than enough to feed people. Are there new unexplored technologies that may be better?

AIM

To conduct research to answer the question: Can genetic engineering biotechnology solve the problem of world hunger?

METHOD

1. As a group, begin by researching genetically modified organisms such as golden rice (and other genetically modified foods produced using biotechnology). Use this researched information and the information provided in Table 8.1 to assist you in making a decision. You may wish to start by using the search phrase, 'the politics of golden rice'.
2. Investigate issues such as:
   - Does this biotechnology pose risks to humans and the environment?
   - Should resources such as this be privatised, with intellectual property rights?
   - Should a small number of companies control much of the seed?
   - Are there other technologies that may be better used to combat poverty and hunger and bring a balance of power and economy?
3. Discuss the issue as a group. Listen with focus as each student expresses his or her own opinion as an evidence-based argument. There is no need for the group to reach consensus.
4. Individual work: Use the results of your research and group discussion to write your own one-page answer to the question posed in the aim of this investigation.
   Using examples, evaluate the potential benefits of researching genetic technologies.
   In your answer:
   a. Identify the issue and the decision to be made.
   b. Describe the technology that is causing the issue, advantages, disadvantages and why it raises moral dilemmas.
   c. Present the facts – what information is needed to make an informed decision about this issue?
   d. Present your opinion, explaining whether your opinion changed over the course of the investigation (and, if so, why). What are the opinions of others in your group?
   e. What other options (alternative outcomes) are there for this issue? Identify both positive outcomes and unacceptable outcomes for this issue.
   f. Make your decision, and close with a statement justifying it.

EXTENSION

Complete the interactive exercise found in the weblink.

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KEY CONCEPTS

- Biotechnologies have social, financial, environmental, legal and ethical impacts.
- Bioethics is the application of ethics to the science and practice of biology.
- The world's population is growing and food shortages may be addressed by using GM food.
- A framework of ethical principles may be applied when making ethical decisions in biology.

CHECK YOUR UNDERSTANDING 8.2

1 Define the following terms:
   a bioethics
   b bioethicist
   c contentious issue.
2 Outline the four basic principles that are used to guide ethical decision making.
3 Outline the advantage of golden rice over normal rice.
4 Explain how golden rice has been genetically engineered.
5 List three contentious issues surrounding the use of golden rice for humanitarian purposes.

8.3 Future directions and benefits of research

Biotechnology is constantly evolving. The techniques and applications used today are far more complex and precise that those used just ten years ago. As new discoveries advance scientific knowledge and understanding, we can predict the impact of future biotechnology on individuals, society and the environment. These new discoveries include advances in biotechnology that have been developed for agriculture, medicine and industry.

Weblink
CRISPR

One of the most recent developments in biotechnology that has enormous implications for the future is a new genome editing technique called **CRISPR** (Clustered Regularly Interspaced Short Palindromic Repeats). The CRISPR enzymes were discovered in bacteria, where their role was to 'chop up' the DNA of invading viruses. CRISPR-Cas 9 is an enzyme that can be used to snip DNA at a particular base, so it can then be attached to a 'guide' RNA (Fig. 8.20) that targets a specific complementary nucleotide sequence in the genome to which it will be added. As a result, genes can now be spliced and inserted with pinpoint accuracy, opening up a host of possibilities, such as uncovering genes that are instrumental in causing neurological disorders such as Alzheimer's disease and schizophrenia. However, the easy use and accuracy of CRISPR also raises concerns about **germline** gene editing and the creation of 'designer babies'.

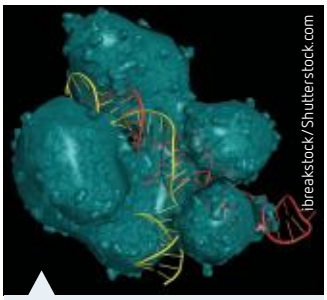

FIGURE 8.20 Graphical representation of CRISPR-Cas9 protein with guide RNA

INVESTIGATION 8.3

Secondary-source investigation of the future direction of biotechnology

Weblink
Boosting wheat fibre

BACKGROUND INFORMATION

The rapid evolution of biotechnological applications is exciting, but also potentially threatening. Genetic technologies may be used for the benefit of humankind, but there is also the possibility of misuse, to the detriment of humans and the environment. Does the threat of abuse of a technology mean we should not explore it further? What are the difficulties in trying to establish and enforce international laws to regulate the use of biotechnologies? »

Weblink
Use of urine
Explore the possibility of extracting nitrogen and phosphorus from urine, for potential use in fertilisers.

Weblink
Sea-quence
Sea-quence is a project to sequence the genomes of nine reef-building corals, together with symbiotic microbes. The information may be used to stop coral loss on the Great Barrier Reef.

AIMS

To research future directions in the use of biotechnology
To evaluate the potential benefits of researching these technologies

METHOD

1 Use secondary sources to research in detail three examples of future directions of the use of biotechnology. Select examples from different fields – agriculture, conservation, medicine and industry.
2 Summarise your findings in a table. Use Table 8.2 as a guide.

TABLE 8.2

TECHNOLOGY	FUTURE DIRECTION FOR USE	POTENTIAL BENEFIT OF RESEARCHING THIS TECHNOLOGY
1		
2		
3		

3 Use an appropriate referencing style to acknowledge your sources.

CONCLUSIONS

Based on the information you have gathered, write a statement in which you evaluate the potential benefits of researching biotechnologies.

You are encouraged to find your own resources, but some are given here in weblinks as a starting point or to generate ideas.

8.4 Changes to Earth's biodiversity

Weblink
Fighting animal extinction
Animal parents 'from the grave' may have their DNA introduced once again into the gene pool.

Sustainability

Ethical understanding

Personal and social capability

Worksheet
Use of modern biotechnology in conservation: White rhinos

Biodiversity is critical in maintaining healthy ecosystems and thereby sustaining plant and animal life on Earth, including human life. Biologists and breeders in agriculture realise they need to conserve diversity in living organisms for the long-term survival of species and to feed the growing human population.

Modern biotechnology gives humans the potential to alter the path of evolution by artificially combining the qualities of organisms that were once separate species (for example, by creating transgenic species). This could increase biodiversity in the short term, by introducing new gene combinations in the population and new genes in individuals. In the long term, however, biodiversity will be reduced if these organisms with desirable characteristics are reproduced and bred, using reproductive technologies such as cloning and selective breeding.

Another concern is that wild varieties of plants and animals may cross-breed with genetically engineered ones, which has the potential to affect Earth's biodiversity.

You have seen in this chapter that biotechnology can be applied to save species that are on the brink of extinction, improve biodiversity in farming practices and alleviate hunger in resource-poor areas of the world, at the same time putting into place biodiversity conservation measures to ensure future resilience in these species.

A major disadvantage of biotechnologies such as selective breeding, cloning and genetic engineering is the potential to reduce genetic diversity in the long term and therefore increase the risk of populations being wiped out in response to disease or a sudden environmental change.

If humans influence the path of evolution to the extent that species become extinct, human survival may also be compromised. A balanced ecosystem with a wide diversity of species is needed.

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The survival of genetically engineered organisms depends on how well they compete with wild-type species in nature. If their genes are advantageous to the population, the frequency of these genes in the gene pool will increase, but the downside of this is that they may out-compete natural species and result in the extinction of those species.

Our ability to apply biotechnology to prepare genetically modified organisms for epidemics by further genetically transforming them with genes for resistance to diseases may prove too costly and time-consuming.

To overcome the threat of reduced diversity, stocks of varied cattle are being maintained, and germplasm of plants and gamete material of animals is being banked, so that a variety of genomes is available if needed in future – for example, in case of unprecedented events.

FIGURE 8.21 Australia's first genetically modified Holstein calves, Holly, Molly, Lolly and Jolly, cloned for their high-protein milk, with Dr Ian Lewis, who collaborated in the project.

INVESTIGATION 8.4

How do genetic techniques affect Earth's biodiversity?

Using information you researched throughout this chapter, produce a multimedia presentation to answer the inquiry question at the start of this chapter: How do genetic techniques affect Earth's biodiversity? Use a range of well-selected images to illustrate your answer.

Work though at least one of the weblinks provided here to further broaden your understanding before completing this task.

Weblink
Genetic technologies
Use this link to work through interactive activities and learn more about genetic technologies, including conservation genetics.

KEY CONCEPTS

- CRISPR is a new gene editing tool that is very precise, and also contentious because it can be used in human gene editing.
- Using CRISPR, genes can be spliced and inserted with pinpoint accuracy.
- Future directions for research include the use of:
  - gene therapy to treat human disease
  - GMOs to alleviate hunger
  - plant banks and animal cryopreservation to maintain biodiversity in agriculture and for wildlife conservation
  - plant- and algae-based resources to develop next-generation biofuels.
- Loss of biodiversity is a major concern, and biotechnology can be used in breeding programs to counteract this.

Weblink
A bigger toolbox: Biotechnology in biodiversity conservation

CHECK YOUR UNDERSTANDING 8.4

1. Outline how the technology known as CRISPR Cas-9 works.
2. List three future directions and benefits of biotechnology.
3. Describe a case study where biotechnology has been used for conservation purposes.

8 CHAPTER SUMMARY

Biotechnology: How do genetic techniques affect Earth's biodiversity?

BIOTECHNOLOGY: USES AND APPLICATIONS

Past (ancient)

- Food production – use of living cells to make bread, cheese and wine
- Medicine
- Crop and animal domestication
- Aboriginal people and aquaculture

Past (classical)

- Fermentation
- Plant and animal selective breeding
- Agriculture
- Medicine and antibiotic production

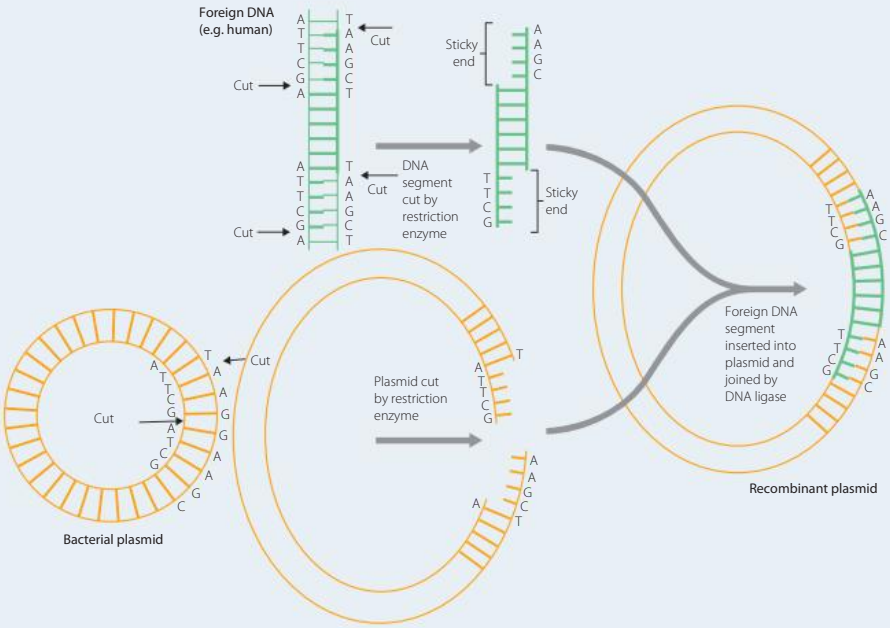
Present (modern)

Technology to manipulate DNA

- DNA splicing
- DNA amplification
- Recombining DNA

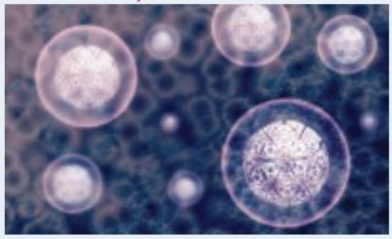
Technology to analyse and visualise DNA

- Gel electrophoresis and gene probes
- DNA sequencing
- DNA profiling



Applications of modern biotechnology

Conservation



Industrial

Agricultural

Medical

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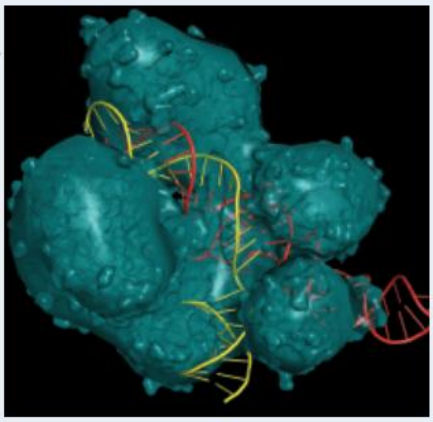
Future directions and potential benefits

Potential benefits to society of research using genetic technologies

Using CRISPR, genes can be spliced and inserted with pinpoint accuracy.



Potential benefits

- Gene therapy to treat human disease
- Genetically modified food to alleviate hunger
- Plant banks and animal cryopreservation to maintain biodiversity and for wildlife conservation
- Plant and algae-based resources to develop next-generation biofuels

Changes to Earth's biodiversity due to genetic techniques

Short term changes:

- Increase biodiversity by introducing new gene combinations.

Long term changes:

- Decrease biodiversity by selectively breeding desired gene combinations. Breeding programs, plant banks are being developed.

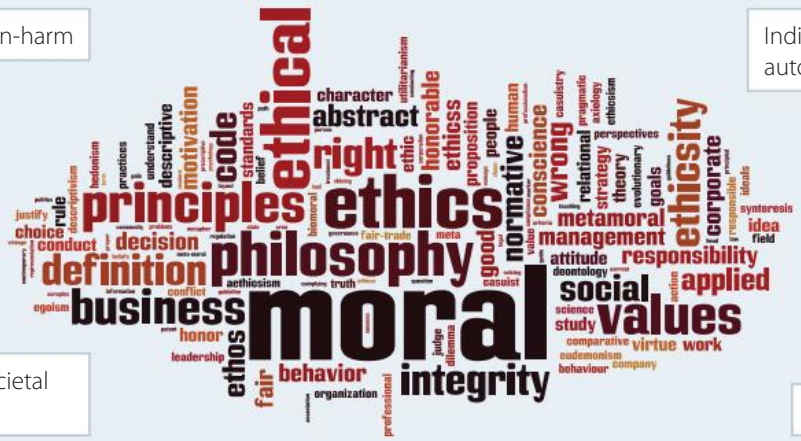
FUTURE BIOTECHNOLOGY

Ethics and social implications (plant and animal uses)

Making an ethical decision

Benefit and non-harm

Individual rights and autonomy



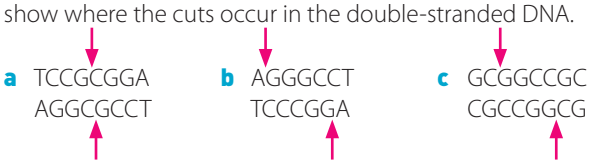
Privacy and societal perception

Equity and justice

8 CHAPTER REVIEW QUESTIONS

Review quiz

1. Define 'biotechnology'.
2. Outline one ancient Aboriginal use of biotechnology.
3. Explain how agriculture began and why breeding of animals with desired characteristics can be described as biotechnology.
4. Outline three biotechnology practices carried out thousands of years ago (other than selective breeding).
5. Why are antibiotics considered to be biotechnology?
6. Discuss how the use of fermentation has evolved as a form of biotechnology.
7. Describe three ways in which genes can be manipulated.
8. Explain the purpose of gel electrophoresis and PCR in gene manipulation.
9. Outline two techniques that can be used to visualise DNA.
10. Distinguish between the use of selective breeding and genetic engineering in creating crops or livestock with greater yields.
11. Explain why seed banks have been established in different places in the world.
12. Outline two ways in which genetic engineering helps conserve endangered animals.
13. Create a flow chart to outline the production of recombinant DNA.
14. Predict whether the following cuts made by restriction enzymes will produce sticky or blunt ends. The arrows show where the cuts occur in the double-stranded DNA.



15. Explain how you could use biotechnology to design and implement a breeding program that minimises inbreeding of an endangered species.
16. Evaluate whether the use of genetically modified food is ethical from a consumer's point of view and from the perspective of alleviating hunger in the world.
17. Answer the inquiry question: How do genetic techniques affect Earth's biodiversity?

Exam preparation

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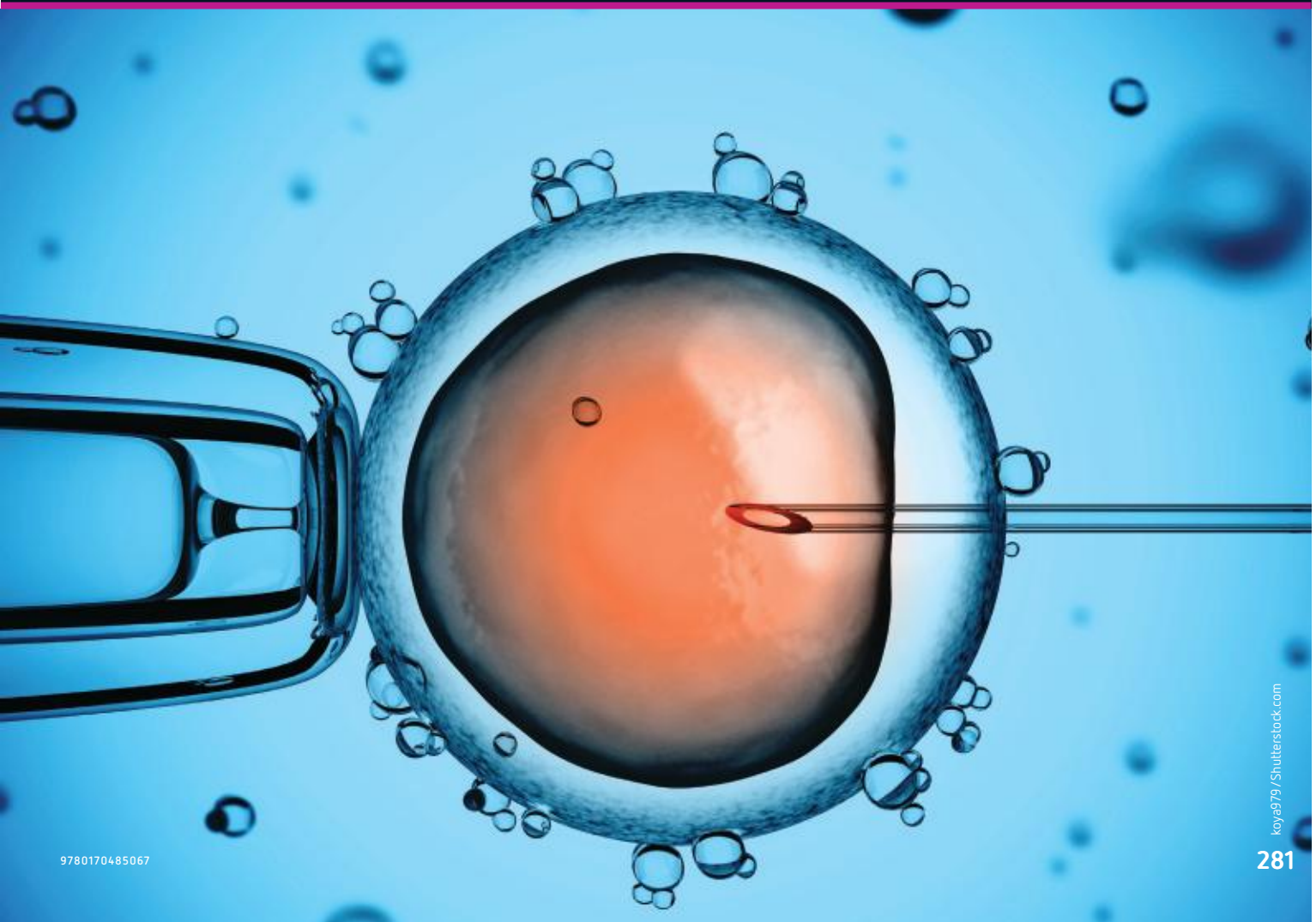
9 Genetic technologies

INQUIRY QUESTION

Does artificial manipulation of DNA have the potential to change populations forever?

Students:

- investigate the uses and advantages of current genetic technologies that induce genetic change
- compare the processes and outcomes of reproductive technologies, including but not limited to: S
  - artificial insemination
  - artificial pollination
- investigate and assess the effectiveness of cloning, including, but not limited to: EU ICT
  - whole organism cloning
  - gene cloning
- describe techniques and applications used in recombinant DNA technology, for example: EU CCT
  - the development of transgenic organisms in agricultural and medical applications (ACSBL087)
- evaluate the benefits of using genetic technologies in agricultural, medical and industrial applications (ACSBL086) S EU
- evaluate the effect on biodiversity of using biotechnology in agriculture S
- interpret a range of secondary sources to assess the influence of social, economic and cultural contexts on a range of biotechnologies EU ICT IU DD



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Worksheet
Genetic technologies

Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
9.1 Modelling and comparing the processes and outcomes of artificial pollination and artificial insemination
9.2 Secondary-source investigation to assess the effectiveness of cloning
9.3 Secondary-source investigation to research technologies that induce genetic change
9.4 Benefits and limitations of genetic technologies: A class debate
9.5 Secondary-source investigation: Effect of biotechnology on biodiversity
9.6 Using secondary sources to assess social, economic and cultural influences on the use of biotechnologies

Worksheets
- Genetic technologies
- Selective breeding in wheat
- Transgenic mammalian animals
- Comparing plant modification methods

Nelson MindTap

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FIGURE 9.1 Scientists are now able to artificially manipulate DNA

For centuries, humans have been able to alter the gene pool through selective breeding, hybridisation, artificial pollination and artificial insemination. With an increased understanding of DNA and gene expression, cloning and recombinant technologies are now possible and scientists can induce desirable genetic changes for the benefit of the human population. This chapter explores some of the processes and techniques used with these technologies and investigates their impacts on society and the environment.

9.1 Processes and outcomes of reproductive technologies

Humans have been trying to breed improved animals and plants for agricultural purposes for thousands of years.

Selective breeding

Selective breeding in animals, in its most basic form, involves mating a male that displays at least one desirable characteristic with a female that displays at least one other desirable characteristic, in the hope that some offspring will inherit the desired favourable genetic traits from both parents. For example, crossing a Friesian bull (female Friesians are known for their production of *large quantities* of milk) with a *Jersey* cow (which produces *creamy* milk) will result in some offspring that produce large amounts of creamy milk. Offspring that have both these desirable traits are then selected for further breeding. In selective breeding, both parent individuals are different varieties of the same species, so the offspring produced are fertile. Selective breeding may have the advantages of hybrid vigour (Chapter 8), but may also introduce some disadvantages. For example, if undesirable genes are inherited together with desirable traits, it is possible that some hybrid cows from a Friesian × Jersey might grow large udders that make walking difficult and cause ulcers where the udder and legs make contact (Fig. 9.2).


UniversalImagesGroup/Getty Images

FIGURE 9.2 Selectively bred cow with very large udder

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Another disadvantage of using whole animals for selective breeding is that it is time-consuming and costly. It involves transporting large animals over long distances. There is always the chance that they will injure each other or that they may not mate, or that it will take a long time for the cow to become pregnant. To overcome these problems, the technologies of artificial insemination, in vitro fertilisation (IVF) and multiple ovulation embryo transfer (MOET) have been introduced.

FIGURE 9.3 A liquid nitrogen bank filled with semen from a chosen bull, to be used to impregnate cows

Artificial insemination

Artificial insemination in animals involves collecting sperm from a chosen male and artificially introducing it into several selected females. There are records of attempting artificial insemination in dogs in the early 1700s but it was only from the 1980s onwards that it became a commercial enterprise and became more widespread. This was due to research into, and discovery of, the effective storage and transport of sperm. Semen containing the sperm is removed from the male (using mechanical stimulation or an artificial vagina), and the collected semen is divided into semen straws, chilled and then frozen in liquid nitrogen for long-term storage and transportation (Fig. 9.3). When it is time to transfer the semen to the female, the semen straw is thawed and placed in a sterile artificial insemination 'gun'. The gun is carefully inserted into the vagina to the cervix, where the semen is deposited.

Sustainability

The outcomes

Transporting frozen sperm overcomes the problem of transporting large animals over long distances, is cost-effective and reduces the danger to animals of injury in transit or during mating. Many females can be inseminated and so one male can sire offspring with several females. Because the semen can be frozen indefinitely, a male can still produce offspring many years after the animal has died. For example, Dutch bull Sunny Boy produced 1.7 million units of semen in the 1990s and offspring were produced for many years after his death. An American stud bull, Toystory, holds the current record. He produced 2.4 million units of semen and has sired 500 000 daughters in 50 countries to date. There are only 50 bulls worldwide who have produced more than a million units of semen. These statistics lead to questions about changing biodiversity.

Artificial insemination is also being used in conservation, to increase the numbers of endangered species. Monash University PhD student Jonathan Daly conducted the first artificial insemination of a shark (using a broad-nosed seven-gill shark as a model species) in trying to develop a technique to apply to grey nurse sharks, whose numbers are dwindling on the east coast of Australia. Mzuri, a male gorilla, was the first gorilla born (in June 1984) using this reproductive technology, which was carried out at Melbourne Zoo by Professor David Galloway of the University of Melbourne (Fig. 9.4).

Artificial insemination can be costly, due to the requirement for specialised equipment, and it is time consuming and has the potential to cause injury to the female if carried out incorrectly. The disadvantage with

FIGURE 9.4 Baby gorilla Mzuri was born as the result of artificial insemination at Melbourne Zoo.

the greatest consequences, however, is the reduction in genetic diversity within populations throughout the world.

The advantages of artificial insemination far outweigh the disadvantages. It has become the principal method of assisted reproductive technology for many types of cattle, sheep and pigs, as well as performance and sport horses around the world.

With the use of selective breeding techniques such as artificial insemination, humans alter the genetic composition of the breeding population, by selecting individuals with traits considered by the breeder to be advantageous, and carrying out controlled breeding of these individuals. As a result, individuals with the desired phenotypes have new combinations of alleles that are introduced into the population and these offspring are in turn selectively bred. The alleles that increase in frequency in the gene pool of the population are therefore those that have been selected by the breeder rather than by nature. An individual's survival and reproduction within the population depend on the presence of alleles that enable them to increase the output of a product that is useful to humans, rather than alleles that increase their fitness in terms of suitability to the environment or reproductive success. Because the selected offspring are fertile, these new combinations of alleles can be passed on to future generations.

In vitro fertilisation

In vitro fertilisation (IVF) differs from artificial insemination in that an egg is fertilised by a sperm outside the mother's body (in an artificially created environment, such as a Petri dish). The resulting zygotes are cultured until they have progressed to an early stage of development. They are then transferred into the biological mother, a surrogate mother or frozen in liquid nitrogen for later transplantation or for use in scientific research. IVF is often carried out in conjunction with MOET to maximise the high genetic merit of female cattle. MOET allows cows, which normally give birth once a year, to become stud breeders when surrogate mothers are used. Direct embryo transfer is the final stage of IVF, where the fresh or frozen cultured embryo is transferred into the uterus. Insertion of the embryo is done using a thin tube called a catheter, which deposits the embryo into the uterine lining.

MOET is multiple ovulation embryo transfer.

This method is often used in cases where there is decreased fertility in one or both of the parents. In vitro fertilisation has the following effects:

1. *Genetic diversity* of populations is reduced due to the production of large numbers of viable embryos from a small selection of parent animals with desirable traits. (See page 291 for more detail.)
2. *Genes for infertility*, which would not naturally have been passed on, are now inherited by offspring. It is important to consider whether humans are breeding infertility into a population by assisting reproduction with reproductive technologies such as in vitro fertilisation. This is the opposite of natural selection, where the frequency of genes that enhance fertility tend to increase.
3. *Sperm banks* have the potential to alter the genetic composition of a population, in animals and in humans. People can choose the sperm donor they prefer, based on a list of his traits. This may increase the frequency of certain donor genes within the population (for example, in humans, academic ability or physically attractive features) and reduce the frequency of those seen as unfavourable. Elimination of certain genes means other important alleles may be lost (such as those associated with creativity or disease resistance).

Artificial pollination

Records show that selective breeding in plants, or **artificial pollination**, was used in ancient times – an Assyrian carving that dates back to 870 BCE shows the artificial pollination of date palms. Artificial pollination was used by Mendel in his experiments with pea plants in the 19th century and was used by scientists, such as William Farrer, who were involved in hybridisation studies; and it is still used today. In the past century, thousands of new breeds of plants have been created using artificial pollination.

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The process

The process of artificial pollination involves removing the stamens of a flower and dusting the pollen onto the stigma of the same flower or another flower on the same plant (self-pollination) or a flower on a different plant (cross-pollination). As with the other selective breeding processes discussed in this chapter, this gives the breeder a greater degree of control over the breeding process.

Artificial hand pollination (Fig. 9.5) is an important horticultural technique that enables the production of offspring with specific favourable characteristics, such as disease-resistant fruit. In order to ensure a greater yield of crop and seeds, humans have artificially pollinated almond crops and date crops for thousands of years.

The process enables the creation of new varieties of plants, but its overuse can lead to crops that are too similar, thus reducing biodiversity. The corn blight that occurred in the USA in 1970 is an example of the impact of a reduction in genetic diversity and increased susceptibility to disease in a crop species.

FIGURE 9.5 Artificial pollination by hand

FIGURE 9.6 Drone pollination of flowers is being explored by scientists.

The outcome

Many crop plants worldwide depend on insect pollination. Insufficient pollination will reduce fruit and seed yield, and can affect the quality of the offspring in both growth rate and resistance to herbivores. Researchers compared artificially pollinated gooseberries with those pollinated by insects. Compared to the hand-pollinated plants, the insect-pollinated plants had larger fruit and greater seed germination rates. As bee populations around the world decline, scientists are currently looking at drone pollinators (Fig. 9.6), which have a sticky surface to take pollen to the flower to ensure direct pollination.

Artificial pollination is often used to produce hybrid plants. The development of maize (corn) has produced a hybrid with an increased growth rate, greater uniformity and increased yield.

Evidence shows that artificial pollination increases genetic variability within populations due to the creation of hybrid species. The use of artificial pollination to create hybrids is an area where genetic diversity increases – *new combinations of alleles* are introduced into the gene pool of a population. For example, wheat hybrids were created by crossing *Purple Straw* variety 14A and a Fife–Indian wheat variety, *Yandilla*, to create a new variety called *Federation*. New gene combinations can be passed on to future generations if the hybrids are fertile, increasing their frequency in the gene pool and thereby altering the genetic composition of the population. (Remember that in hybridisation *within a species*, the resulting hybrids are *fertile*, as opposed to hybridisation *across species*, where the offspring are usually infertile.)

Worksheet
Selective breeding in wheat

INVESTIGATION 9.1

Modelling and comparing the processes and outcomes of artificial pollination and artificial insemination

Weblink
Artificial pollination

Weblink
Artificial insemination

In this investigation, you will use a model to illustrate the differences in the processes used and the outcomes of reproductive techniques. A model is a visual or physical representation (Chapter 1, page 5) and can include illustrations and diagrams, a three-dimensional model or a digital model.

METHOD

Select or create a model to illustrate the processes used for each reproductive technique example and the outcome of the process.

Ensure that the model is labelled so that it clearly compares the processes and outcomes of each of the reproductive techniques.

See the weblinks as a resource for this activity.

RESULTS

Ensure that your model is appropriately labelled or coded and that the similarities and differences between the processes and outcomes are clearly shown.

Models in biology and their purpose are dealt with in more detail in Chapter 1, page 5.

PEER REVIEW OF THE MODEL

Use the following template to evaluate another student's work by indicating positive aspects as 'warm' feedback and aspects that don't match the criteria as 'cool' feedback. Use the feedback to design improvements to your own model.

	PEER FEEDBACK
Warm feedback	
Cool feedback	

CONCLUSION

Evaluate your model. Consider the feedback from the peer review exercise: are there any improvements to the model that should be made?

KEY CONCEPTS

- Artificial insemination involves the process of inserting semen into the vagina of an animal. It allows for animals with desired characteristics to produce offspring.
- Artificial selection is a process that has enabled the selection of specific characteristics in animals for the benefit of humans.
- Artificial pollination involves the dusting of pollen by hand, from the anthers to the stigma of the same flower or a different flower. This enables the plant breeder to control the characteristics of the plants being bred.
- Artificial pollination has been used to produce plants with specific traits that are beneficial for humans.
- The aim of reproductive technologies is to pass on desirable characteristics to the next generation.

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CHECK YOUR UNDERSTANDING 9.1

1 Define the following reproductive technologies and provide an example of each.
   a selective breeding
   b artificial insemination
   c in vitro fertilisation
   d artificial pollination
2 List three outcomes of the reproductive technologies named in Question 1.
3 State the main difference between *within-species* hybridisation and *between-species* hybridisation.

9.2 Cloning

Selective breeding as described in the previous section relies to some extent on trial and error – hoping that the desired combination of favourable genes ends up in some individuals. A modern-day technology that overcomes the trial-and-error nature of selective breeding is cloning. **Cloning** is the production of an exact copy.

Two forms of cloning are discussed in this chapter.

- **Gene cloning** occurs at a *cellular level* and involves producing identical copies of one *gene*. Multiple copies of a gene are needed for genetic engineering and biotechnological research.
- **Whole-organism cloning**, also known as **reproductive cloning**, involves creating a genetically identical (whole) organism, using a somatic cell (or a few somatic cells) from another mature organism. *Whole-organism* cloning is a form of asexual reproduction and so it is considered a **reproductive technology**.

Other forms of cloning include cell cloning, which is used for unicellular organisms, and molecular cloning, which is used in recombinant DNA technologies.

Gene cloning

In gene cloning, scientists select a gene, remove it from the source DNA and insert it into the DNA of another organism, to make identical copies of that gene. This technique is used in the production of insulin on a large scale.

The simplified steps involved in the process of gene cloning are outlined below and in Figure 9.7.

1 The gene (section of DNA) is cut from the source organism using restriction enzymes (enzymes produced by bacteria).
2 The gene is pasted into a vector DNA or plasmid by a process known as ligation (ligase enzymes are used to join fragments of DNA).
3 The plasmid containing the gene is introduced to a host cell by a process called *transformation*.
4 The host cell can now make copies of the vector DNA when it makes copies of its own DNA.

Polymerase chain reaction (PCR) is a form of *in vitro* DNA cloning (carried out in a test tube rather than a living organism). PCR is used widely in research and has many genetic applications. It amplifies a particular DNA sequence and makes multiple copies that can then be used in various research and analysis techniques. PCR involves a process of thermal cycling to denature the DNA strand and the use of complementary primers that locate and duplicate the required section of DNA. The three processes are *denaturing*, *annealing* and *extension* (Fig. 9.8) to make multiple copies of the segment of DNA.

FIGURE 9.7 The process of gene cloning

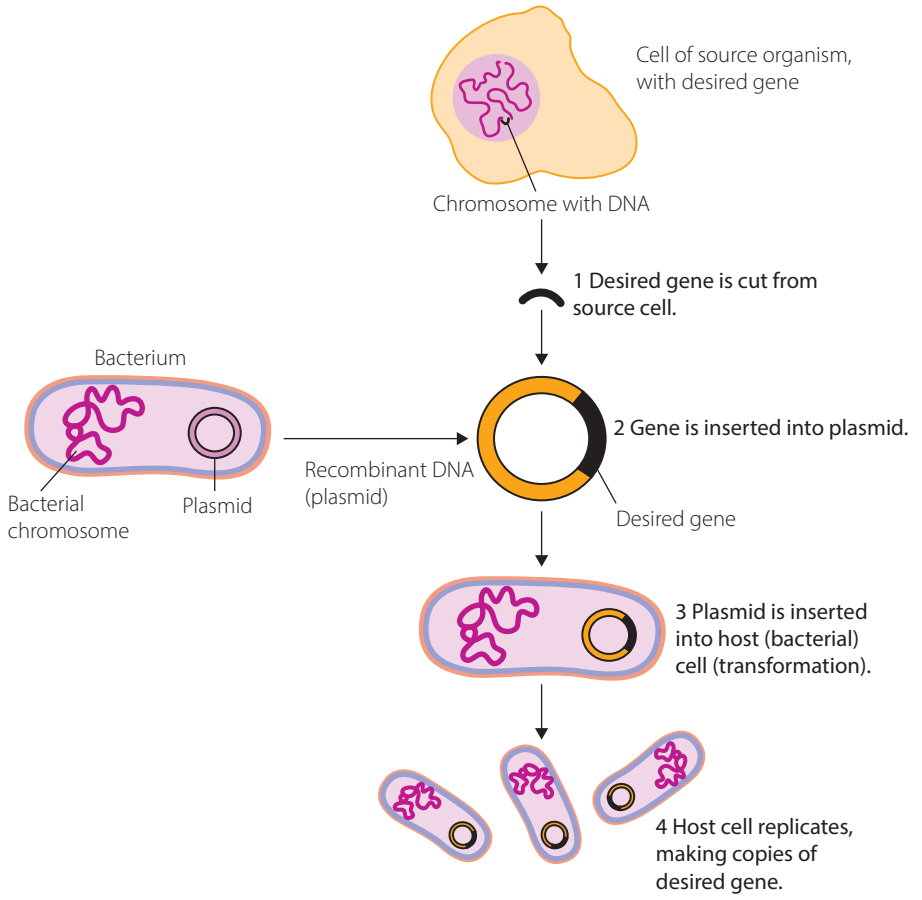
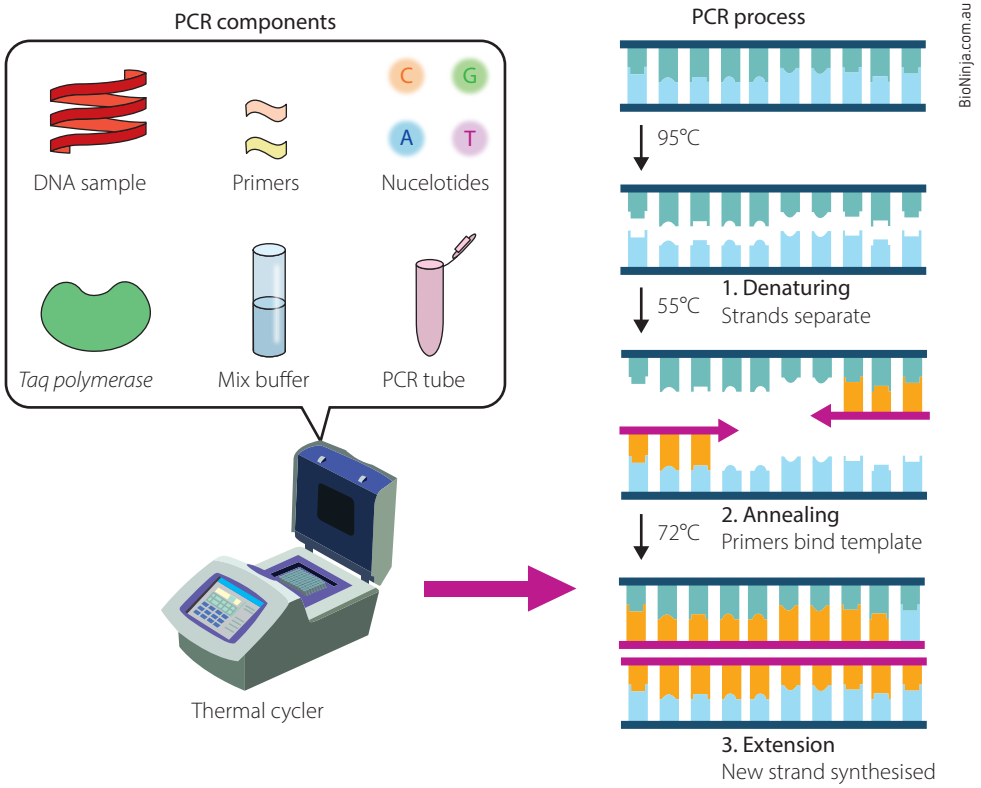


FIGURE 9.8 Polymerase chain reaction



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Whole-organism cloning

The first mammal to be artificially cloned from an adult cell was a sheep named Dolly, born on 5 July 1996 at the Roslin Institute in Edinburgh, Scotland. Dolly was cloned from mature cells taken from the udder of a 6-year-old sheep. It is the *age* of the parent animal from which modern-day clones are produced that makes this process so remarkable. Embryo-splitting leading to multiple births occurs naturally and can be fairly easily replicated in a laboratory. However, cloning from *adult cells* (of living or even deceased organisms) does not occur in nature. The main difficulty faced by scientists who produce clones from adult cells is that certain genes, especially those needed for development, have been 'switched off' or shut down in differentiated cells and so they need to be 'switched on' or reprogrammed for development.

Cloning techniques

The methodology for cloning was tested, proven and patented in 1996. It took about 276 attempts before the success of Dolly the sheep. The rate of success, although improving, is still low. This makes cloning a very expensive technology. Since the start of the 21st century, many mammals have been cloned, including *Tetra*, a rhesus macaque born in 2000 and the first primate cloned (Fig. 9.9). The first cloned horse, *Prometea*, was born in Italy in 2003. The international rules of horse racing do not permit artificial insemination or fertility treatment for the breeding of horses, so it is unlikely that cloning will be acceptable in horse racing in the near future.

Cloning is most commonly used in agriculture. For example, beef from cloned cattle is available in supermarkets in Japan, and seedless grapes, consumed worldwide, are a product of plant cloning.

FIGURE 9.9 Tetra, the first cloned primate, was cloned via embryo splitting.

Somatic cell nuclear transfer

Whole-organism cloning (reproductive cloning) is also known as somatic cell nuclear transfer (SCNT). Each time a mammal is cloned, the SCNT process involves three animals: one that donates the nucleus, one that acts as an egg donor and one that plays the role of surrogate mother.

Ian Wilmut and his team used the method of SCNT described below to create Dolly the sheep (Fig. 9.10).

1. Cells were taken from the udder (mammary glands) of a six-year-old ewe (*sheep number 1*). These cells were starved of nutrients to stop them dividing.
2. The nucleus was removed from a healthy unfertilised egg from another sheep (*sheep number 2*), a process called enucleation.
3. The udder cell with a nucleus, taken from sheep 1 was injected into the enucleated egg of sheep 2. The two cells were then treated with electricity, which caused the cells to fuse or blend together to form a 'fertilised' egg cell. This cell underwent normal growth and development, dividing by the process of mitosis. As the cells continued to divide, the resulting 'embryo' was implanted into the uterus of a third sheep (*sheep number 3*), who was the surrogate mother. The embryo developed and was born as a genetically identical twin to sheep 1, the original sheep that donated the cell from its udder.

Weblink
Whole-organism cloning

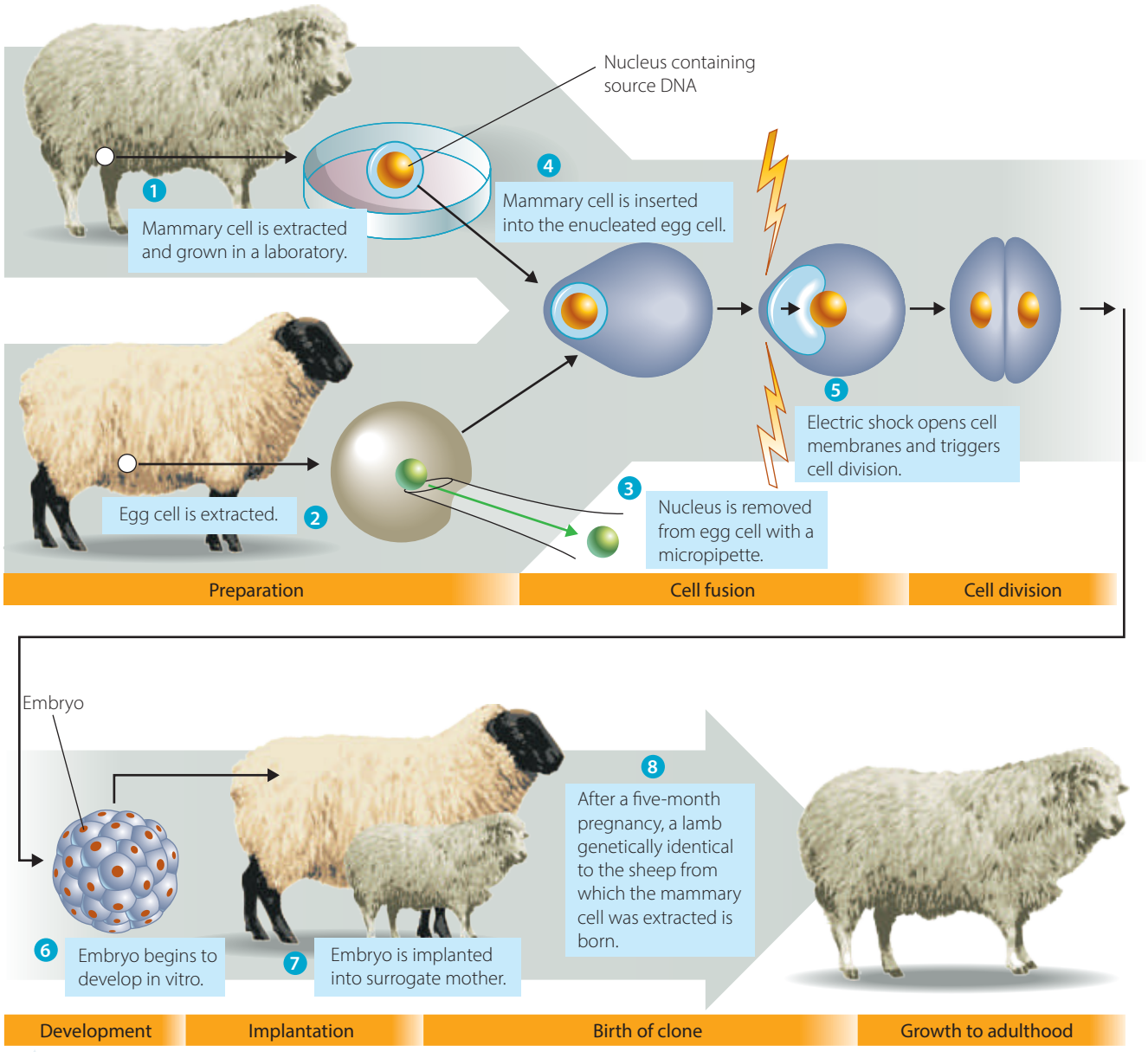

FIGURE 9.10 The process of somatic cell nuclear transfer in sheep

A second technique used in whole-organism cloning is artificial embryo twinning. This is a relatively inexpensive technique: a short time after fertilisation, the embryo is split into two before the cells become specialised. Splitting is done in the laboratory and the embryos are then implanted into a surrogate to develop. The embryos that develop are genetically identical. This technique is used extensively in cattle production.

In horticulture, the cloning of plants is common practice and plant propagation has a long history. Plant propagation involves using a cutting of an existing plant and growing it to form a new plant. Both plants are genetically identical. The propagation of grape vines is a practice that dates back to early European civilisation. The technique conserves the variety and is often faster than growing another plant from seed.

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Tissue culture is another cloning technique used in horticulture. Small portions of plant tissue are grown in solid growth medium or nutrient agar broth until they are ready to be transplanted. The technique is called micropropagation.

Cloning: a contentious issue

Cloning is not a new phenomenon – identical twins are naturally produced genetic clones and occur in every animal species studied for this phenomenon. Cloning is also common in *plants* – botanists have been using cuttings, the splitting of bulbs and other forms of asexual reproduction to propagate plants for many centuries. Bananas are also clones of each other and vastly different from their ancestral stock. They have a very limited gene pool. *Animals* were first cloned a century ago, when two-celled embryos of sea urchins and salamanders were split.

When assessing ethical issues in cloning, you need to consider which type of cloning is being assessed, as the processes vary. Ethical issues are often centred around whole-organism and human cloning rather than gene or cell cloning.

Some of the ethical issues involved in cloning include the following:

Ethical understanding

- Concern for animal welfare – there are already concerns about the treatment of animals in large-scale farming, which many feel would be exacerbated by cloning animals for food production.
- The same techniques used to clone animals could be used to clone humans. This raises moral concerns as well as religious and legal concerns.
- There is the religious argument that, in cloning animals, humans are 'acting as God'.
- There are unforeseen health risks for cloned animals that are yet to be considered.
- Reproductive cloning is an expensive procedure, which limits access to this technique.

Cloning alters the genetic composition of a population

In agriculture, cloning is used as a form of selective breeding once an 'ideal' hybrid has been obtained. The advantage of cloning is that it reduces the 'unknown' element in selective breeding – the characteristics being bred can be precisely controlled. It also allows the desired characteristic to be reproduced in a short space of time, making it an efficient method of obtaining desired characteristics in organisms.

However, cloning reduces genetic diversity in populations, as organisms produced by reproductive cloning are derived from only one parent (an artificial form of asexual reproduction) and are genetically identical to the parent. Only a few parent animals may be cloned, so if many clones are produced from only a few parent organisms, genetic diversity is reduced, as all the organisms have identical DNA. After continued selective breeding, the cloned organism becomes the predominant organism. Cloning ensures that identical combinations of genes (that is, entire genomes) are conserved within a population. It therefore increases the frequency of these genotypes within the population and, as a result of artificial selection, natural gene combinations that are not selected will gradually disappear.

In nature, genes are conserved by evolution only if they serve an essential function for the organism. For example, the genes for cytochromes (proteins essential for chemical respiration) are conserved because, if they are altered by mutation, the individual cannot survive. Therefore, a mutation in these genes that altered the proteins would not be passed on to future generations. The disadvantage of cloning is that, if all members of a species are identical, the population is less likely to survive any sudden environmental changes and is vulnerable to foreign pathogens.

INVESTIGATION 9.2

Secondary-source investigation to assess the effectiveness of cloning

Information and communication technology capability

Ethical understanding

In this secondary-source investigation, you will assess the effectiveness of different cloning techniques and consider the ethical impacts of the development of cloning techniques.

AIM

Write an aim for this investigation. What is the main purpose of this investigation?

METHOD

Use key terms to complete an Internet search and locate current examples of gene cloning and whole-organism cloning. The examples in this chapter can be used as a guide.

Develop a set of criteria and construct a table similar to the one below to collect the data.

WHOLE ORGANISM OR GENE CLONING?	EXAMPLE	DOES IT ACHIEVE THE DESIRED OUTCOME?	ARE THERE ANY LIMITATIONS?	IS IT COST EFFECTIVE?	WHAT ARE THE ETHICAL CONSIDERATIONS?	SOURCE

Use the weblinks as a starting point.

Weblink
Learn genetics: cloning, and click and clone

DISCUSSION

Discuss these questions as a class before writing your answers.

1. Of the examples investigated, which is the most effective? Justify your answer.
2. Which techniques are affected by ethical considerations? Give specific examples.
3. What factors did you consider when you were deciding on the effectiveness of each technique?

CONCLUSION

Write a conclusion that assesses the effectiveness of cloning.

KEY CONCEPTS

- Cloning involves the production of an individual that is genetically identical to one that already exists.
- Two types of cloning are: whole-organism cloning and gene cloning.
- Whole-organism cloning (reproductive cloning) is used to create a genetically identical whole organism.
- Gene cloning is used to produce identical copies of a specific gene.
- Cloning can be used as a form of selective breeding to produce organisms with specific desired characteristics.
- A disadvantage of cloning is that all members of the cloned population are genetically identical and therefore the population is susceptible to specific selecting agents.

CHECK YOUR UNDERSTANDING 9.2

1. Distinguish between whole-organism cloning and gene cloning.
2. Outline a process used to clone whole organisms.
3. Explain the purpose of whole-organism cloning.
4. Explain why Dolly the sheep was a genetically identical twin, not the daughter, of sheep 1.
5. Describe a process used to clone genes.

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9.3 Recombinant DNA technologies

The aim of recombinant DNA technology is to insert a gene from one species into the genome of another. The process involves isolating DNA fragments from one genome using **restriction enzymes**, which can cut DNA at a precise sequence of bases. These can be reassembled using a process known as **ligation**. DNA pieces are joined using an enzyme called DNA ligase. The resulting DNA fragment is called **recombinant DNA**, as it has been recombined with DNA from other sources.

The process of recombinant DNA technology is shown in Figure 9.11. The steps are as follows:

1. The required gene is isolated from a cell.
2. A piece of circular DNA known as a plasmid is removed from bacteria.
3. Two pieces of DNA are cut using the same restriction enzyme.
4. The fragments produced have matching **sticky ends** (sections of single-stranded DNA with exposed nucleotide bases at the end of a double-stranded molecule; Fig 9.12).
5. The bacterial plasmid is cut at two points using the same restriction enzymes.
6. As the sticky ends of the human gene and the plasmid come together, they can join up via base pairing. This process is called annealing.
7. DNA fragments are joined by the enzyme DNA ligase. Joined fragments can form a circular plasmid or a linear molecule.
8. The plasmid is inserted back into a bacterial cell, where multiple copies of the gene can be produced.

Once multiple copies of the gene have been produced, the gene can be inserted into an egg cell of another species and, after fertilisation, becomes part of the newly formed organism's DNA.

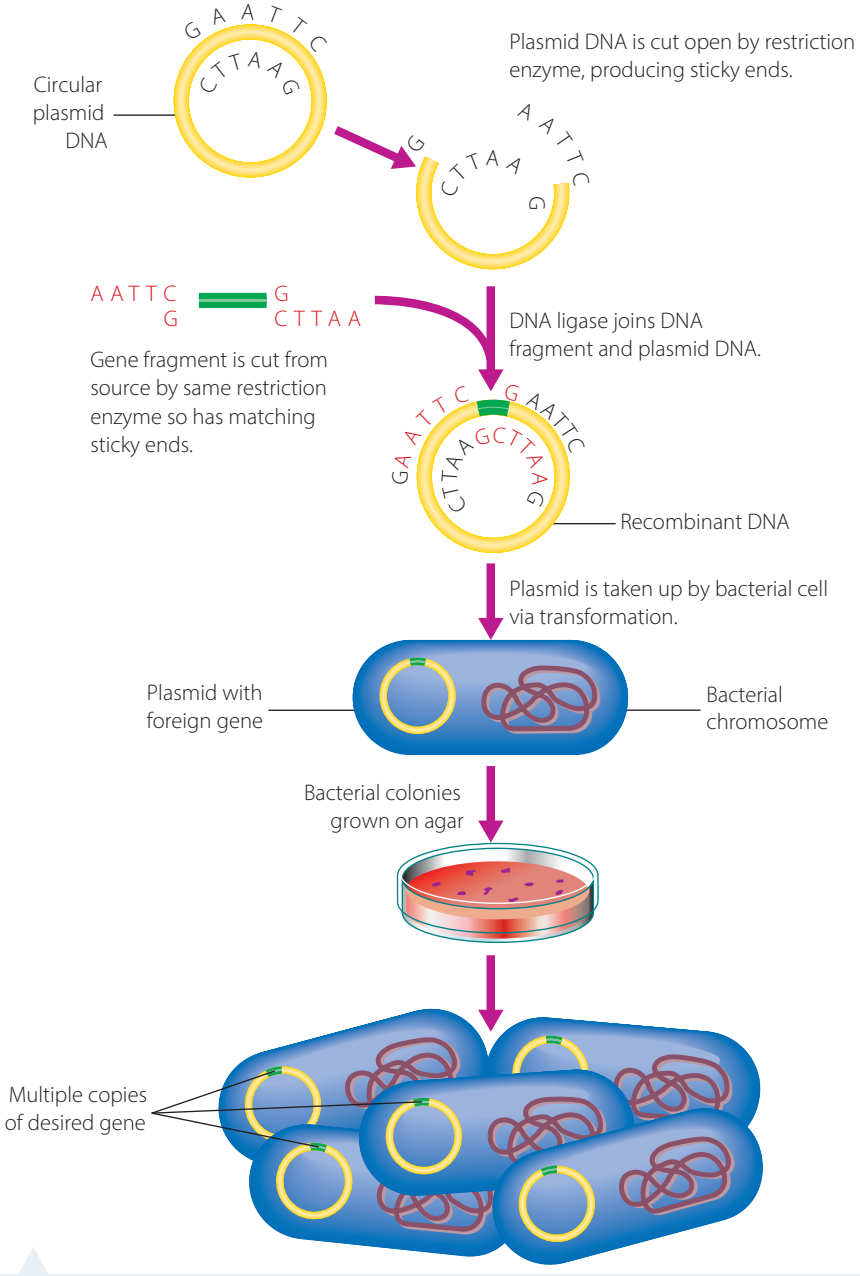

FIGURE 9.11 Recombinant DNA technology

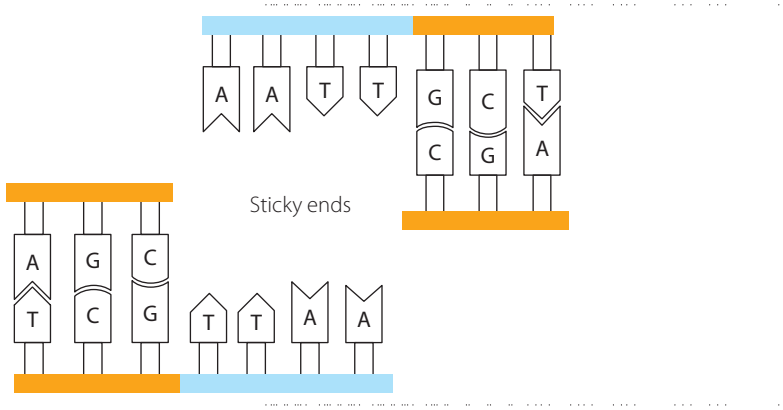

FIGURE 9.12 Sticky ends of a piece of DNA after being cut by a restriction enzyme

Delivering the gene

There are four main ways of inserting the desired gene into the genome of a species to be genetically transformed:

1. **micro-injection** of DNA directly into the nucleus of a single cell – this is usually done under an optical microscope to introduce DNA into egg cells when creating transgenic species (Fig. 9.13a)
2. **biolistics** – mechanically delivering DNA on microscopic particles into target tissues and cells by 'firing' them from a gene 'gun'; for example, tiny gold particles are used to coat the DNA, which is then fired at the target cells under high pressure or voltage
3. **electroporation** – increasing the membrane permeability by applying an electrical current
4. **transduction** by a vector – DNA is carried into cells by a *viral vector* such as an adenovirus, liposome or bacterial plasmid. These vectors may be injected directly into the bloodstream or may be delivered by aerosol delivery (nasal spray or oral aerosol for example, used in trials of gene therapy for cystic fibrosis) (Fig. 9.13b).

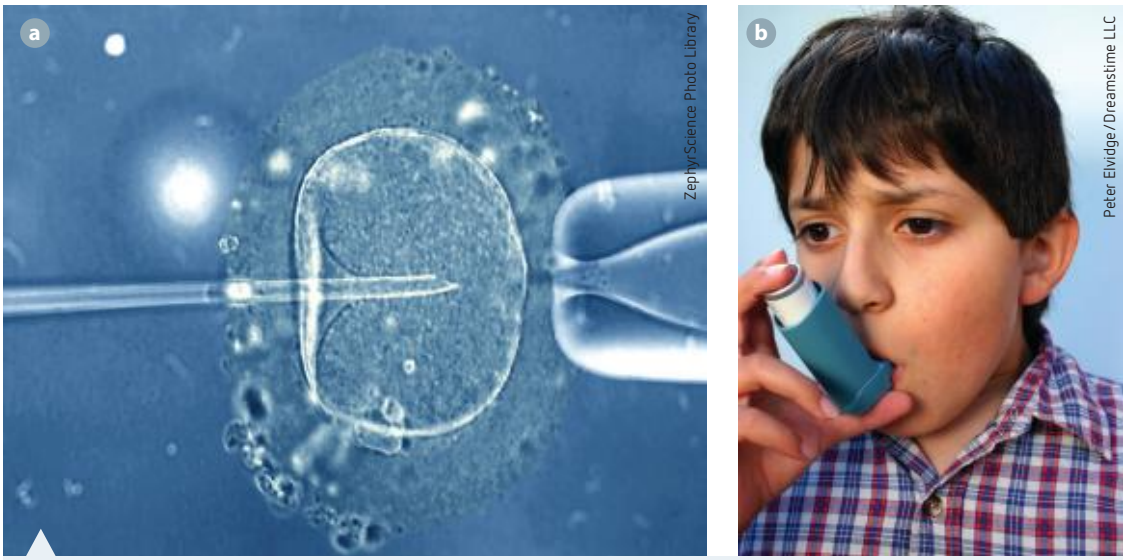

FIGURE 9.13 Two methods of inserting genes into the cells of a different species: **a** micro-injection of DNA directly into the nucleus of a cell; **b** inhaling viral vectors in an aerosol

In **gene therapy,** a healthy copy of a gene is inserted into defective non-germline tissue in a developed or mature plant or animal. Because the gene is inserted into *non-germline* tissue only, it will not be passed on to the next generation. Gene therapy is a new form of medicine, with the potential to replace conventional treatments for diseases.

Transgenic species

Gene manipulation, known as **genetic engineering**, can be used to create transgenic species. A **transgenic species** (*trans* = across) is one that has been created by moving a gene 'across' species – taking a gene from one species and inserting it into the DNA of another species. This gene must become part of that organism's germline genome (DNA) if it is to be inherited by subsequent generations. The gene is therefore inserted either directly into a germline cell or into a fertilised egg cell. The creation of transgenic species is only considered to be a reproductive technology if it increases the reproductive capacity of an animal. Most transgenic species are created for reasons other than increasing reproduction.

Worksheet
Transgenic mammalian animals

Transgenic Bt cotton plants

Over the years, traditional pesticides used on cotton plants had to be made stronger and be applied more frequently to eradicate insect pests. One such insect pest was the caterpillar of the *Helicoverpa zea* moth, which destroys hundreds of millions of dollars' worth of cotton plants each year. With increased spraying, these caterpillars were developing resistance to the pesticides due to the process of natural selection.

In the 1990s, CSIRO scientists in collaboration with the US company Monsanto extensively trialled the use of its *Ingard GM cotton*, also known as **Bt cotton**, which is a transgenic species.

Bt cotton plants were genetically modified (this means to add or remove genes) – they contain a gene that codes for the production of a protein that kills the caterpillar of the *Helicoverpa zea* moth. The insertion of the Bt gene into the cotton plant has reduced the need to use pesticides to kill these caterpillars. This is better for the environment and reduces the development of pesticide resistance in the caterpillars. The gene is called Bt because it was originally taken from the soil bacterium, *Bacillus thuringiensis*.

Cotton growers in New South Wales and Queensland would normally spray their crops numerous times in one growing season. They now only spray occasionally using a narrow-spectrum pesticide to eliminate sucking insects and mites. This spray does not kill beneficial insects (such as ladybirds and wasps), unlike broad-spectrum sprays.

The Bt gene codes for the production of the toxic protein in an inactive form that is harmless to humans and most animals, and even to most insects. However, when the protein is eaten by a caterpillar, it is converted by the caterpillar's digestive system into an active form that kills the caterpillar.

The process used to produce transgenic cotton is as follows:

1. Scientists cut normal cotton seedlings into small pieces and place them on a solid growth medium, where they grow into calluses (Fig. 9.14). After about six weeks the callus cells are transferred to a liquid medium, where they are given hormones to induce them to grow into cotton plant embryos.
2. By genetic engineering, the Bt gene is *cut* from the genome of the bacterium, *Bacillus thuringiensis*, using restriction enzymes.
3. The Bt gene is transferred to the cotton plant embryos. This is done using a second bacterium as a carrier or vector. This bacterium, called *Agrobacterium tumefaciens*, causes crown gall disease in plants, and is used as a vector because it is able to inject genes into other cells.
4. The cotton plant embryos are dipped in a solution that contains a mixture of the vector, *Agrobacterium*, and the extracted Bt genes (which have recombined with the *Agrobacterium* genome), and the vector bacteria inject the Bt genes into the cotton cells.
5. Once the gene is inserted, the embryos containing the Bt genes are grown in tissue culture and are then placed on another solid medium and germinated into small plants, which are planted in pots and grown in glasshouses. These plants are now a transgenic species, containing a gene from another species in their genome.

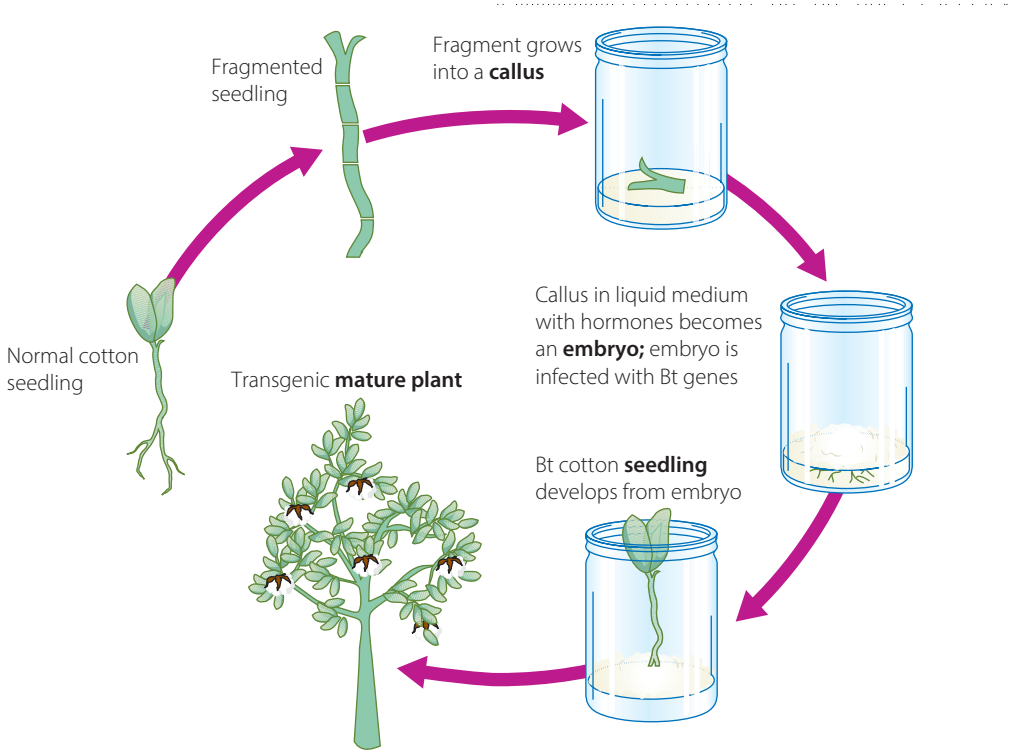

FIGURE 9.14 The process of producing Bt cotton plants by tissue culturing

Cotton is a very difficult plant to culture, but CSIRO scientists have developed workable techniques for growing three varieties of cotton. In order to increase the success of the project, the scientists have developed four different insecticidal genes to use in cotton. However, the project has been controversial, with critics claiming that it is harming the surrounding ecosystems.

Ingard, a cotton containing the product of a single gene, has been replaced by *Bollgard II* cotton, which contains two inserted genes and produces two proteins that are lethal to the caterpillar. It is highly unlikely that the caterpillar will become resistant to both proteins. In addition, cotton farmers plant a 'refuge crop' of pea plants in a field nearby, so that moths that may have one recessive allele for resistance to *Bollgard II* continue to interbreed with moths who feed on the 'refuge crop'. This reduces the chances of inbreeding caterpillars with double-recessive alleles, which could confer resistance.

Another example of a transgenic species is the alfalfa plant in Australia, which has been genetically modified to produce high levels of cysteine. Sheep that graze on this alfalfa have higher-quality wool. Researchers are trying to develop a method to insert this gene directly into sheep.

In the short term, creating transgenic species increases genetic diversity – genes are moved from one species to another and this can be used to confer resistance on species that previously were susceptible to particular diseases, allowing them to survive and pass on their favourable genes. However, in the long term, it may reduce genetic diversity, because the original genetic material of some organisms may be reduced or lost forever – that is, there may be loss of biodiversity.

Medical uses of transgenic organisms

Although there are many ethical issues concerning the use of transgenic organisms in medical applications, their use can expand our understanding of the role genes play in the development of diseases. Transgenic organisms also provide us with opportunities to safely test and develop new treatments. While transgenic animals are not commonly used on the open market as a food source, there are several examples of transgenic animals being used in medical applications. Transgenic sheep in Australia have been given a gene for the blood-clotting factor that is lacking in people who suffer from haemophilia. The factor can be extracted from the sheep's milk and used for human treatment.

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Transgenic animals can be used to study how genes regulate specific body functions. Mario R. Capecchi, at the University of Utah, won the 2007 Nobel Prize in Physiology or Medicine for his work on transgenic mice, in particular, the 'knock-out' mouse. A knock-out mouse is one in which researchers have inactivated, or 'knocked out', an existing gene. This knocked-out gene has been replaced with an artificial piece of DNA at the blastocyst stage of development (Fig. 9.15). Transgenic mice with gonads that have knock-out stem cells are cross-bred, producing some offspring that are pure breeding for the knockout gene. These are used for research. By making a specific gene inactive, and observing differences from normal behaviour or functioning, researchers can infer the function of that gene.

Transgenic mice are often used to study a range of diseases including cancer, obesity, heart disease, diabetes, Alzheimer's disease and Parkinson's disease. Mice are a useful model in medical research because their tissues and organs are similar to those of humans and therefore many of their genes are also similar. Knock-out mice that lack the tumour suppression gene are useful in human cancer research, enabling cancer treatments such as potential drugs, as well as symptoms of the disease, to be studied.

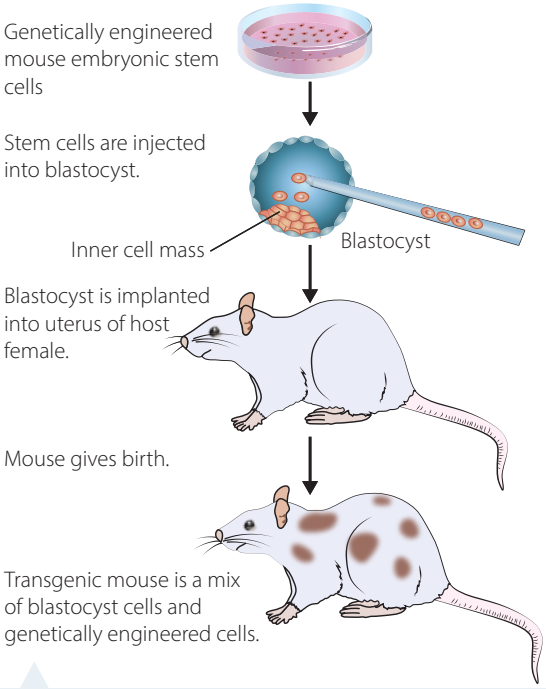


FIGURE 9.15 Producing knock-out mice

In vaccine research, the production of recombinant DNA vaccines involves selecting a gene that codes for a particular antigen, rather than using the whole genome of the antigen. In the case of the recombined vaccine against the hepatitis B virus, the target DNA is inserted into a plasmid that is recombined with a yeast cell (Fig. 9.16). This is incubated to allow the yeast cells to multiply. The antigen is extracted and purified as a vaccine. Vaccine research using recombined DNA is currently being used to develop vaccines against malaria and cattle tick. The advantages of recombinant vaccines are that they have a low risk of side effects and are relatively cheap to produce, making them a good option for use in developing countries.

Weblink
Transgenic mice

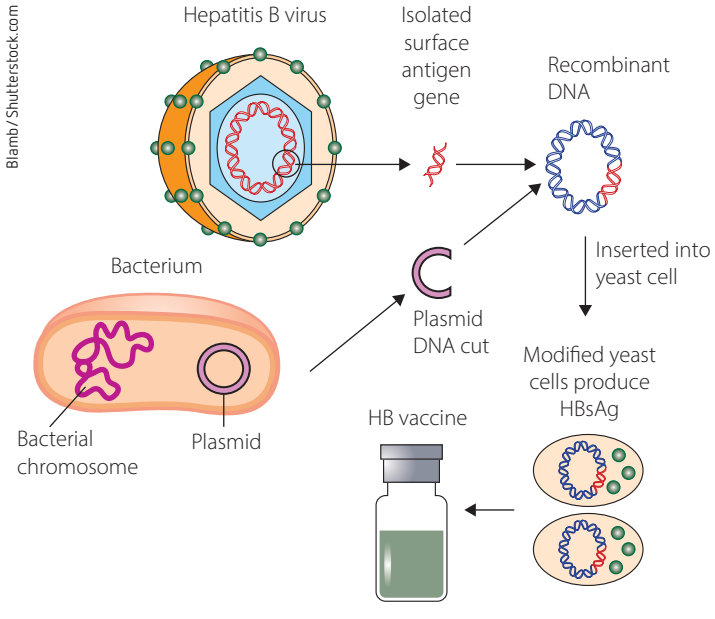
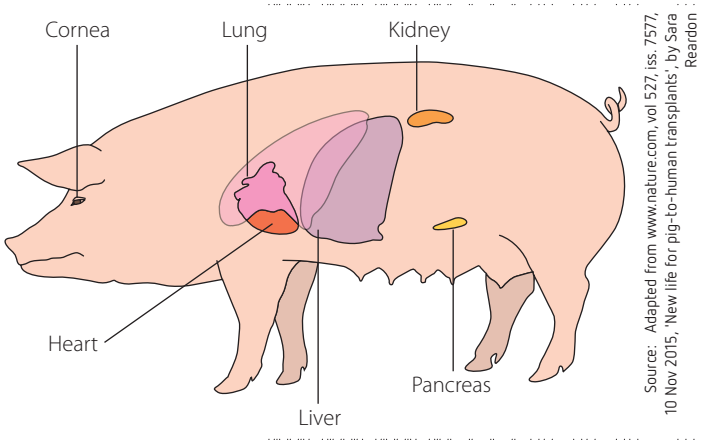


FIGURE 9.16 Production of hepatitis B vaccine

As the demand for human organ transplants grows, there is a need to develop non-human organs for transplantation, as the waiting lists for viable organs that match can be very long. **Xenotransplantation** (using organs from other animals) provides the opportunity for patients with failing organs to have transplants. Organs from transgenic swine that have human complementary **surface markers** (proteins on the cell surface that identify the cell lineage and stage of differentiation) on their cells would inhibit the activation of organ rejection. This would enable people on long waiting lists to access viable organs (Fig. 9.17).

FIGURE 9.17 Potential transplant organs from genetically modified pigs



Source: Adapted from www.nature.com, vol 527, iss. 7577, 10 Nov 2015, 'New life for pig-to-human transplants', by Sara Reardon

KEY CONCEPTS

- Recombinant DNA technology makes it possible to insert a gene from one species into the genome of another species.
- Restriction enzymes are used to cut DNA at a precise sequence of bases, leaving a sticky end.
- Sticky ends can join up to the sticky ends of a bacterial plasmid DNA cut with the same restriction enzyme and joined using an enzyme called DNA ligase.
- The four main ways of inserting the desired gene into the genome of a species to be genetically transformed are: micro-injection, biolistics, electroporation and transduction.
- A transgenic species is one that has been created by moving a gene 'across' species.
- Transgenic species are useful in agriculture and medical research.

CHECK YOUR UNDERSTANDING 9.3

1. Define the following terms: recombinant DNA, sticky end, transgenic organism, vector, plasmid, genetic engineering.
2. Use an example to outline how gene therapy can be used to treat a medical condition.
3. Use a diagram to explain how a transgenic organism can be created.
4. Outline a recombinant DNA procedure used to produce a transgenic organism in medicine and in agriculture.

9.4 Current genetic technologies that induce genetic change

A range of current genetic technologies have been discussed in this and the previous chapter, including hybridisation, selective breeding, artificial pollination, artificial insemination, transgenic species, gene cloning, whole-organism cloning and recombinant DNA techniques. All these techniques produce genetic change, and each has a particular purpose and application.

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INVESTIGATION 9.3

Secondary-source investigation to research technologies that induce genetic change

AIM

To investigate the uses and advantages of current technologies that induce genetic change

METHOD

1. Choose one of the following technologies to study in depth:
   - Hybridisation
   - Selective breeding
   - Artificial pollination
   - Artificial insemination
   - Transgenic species
   - Gene cloning
   - Whole-organism cloning.
2. Use secondary sources to find a range of applications for the chosen technology. Assess the reliability of each source you use.

RESULTS

1. List the advantages of using this technology. Advantages could include financial benefits, health benefits or sustainability of food supplies.
2. Choose a suitable medium to construct an advertisement that promotes the use of your chosen technology to the general population. Your advertisement should show the technology process in a way that can be readily understood and highlight the positive aspects of the technology for a particular application.

Weblink
Mechanisms of recombination

Weblink
DNA cutting and pasting

Weblink
Gene therapy

Weblink
Gene splicing

Weblink
Knock-out mice fact sheet

9.5 Benefits of using genetic technologies

The benefits of genetic technologies to society include improved food quality, cost-effective production and improved food supply. The ability to supply sustainable and nutritional food sources for a growing world population is essential. Improved access to pharmaceuticals and improved techniques in medical research are certainly beneficial to the human population.

Agricultural benefits

Proponents of transgenic organisms suggest that, with gene technology, it is possible to produce crop and animal varieties that are better suited to specific environments, such as high salinity or drought. Plants can be made pest resistant (for example, Bt cotton). For producers, the use of transgenic organisms provides an opportunity to increase the productivity of marginalised land and reduce post-harvest losses.

There have been significant advances in the production of genetically modified plants that have enhanced nutrient levels. For example, rice can be produced with a higher protein or iron content. Increasing the lipid content of starch-rich plants can improve the nutritional value or energy density of crops. A higher lipid content in plants can also be useful in the production of animal feed, fuel and oil, and in industrial applications.

A range of GM animals have the potential to be used as food sources. One of the first GM animals to be approved for human consumption is the GM Atlantic salmon, which is available on the open market and approved in the USA. It has been modified to grow faster and larger than the natural stock by incorporating DNA from Chinook salmon.

Medical benefits

The study of human genomics involves the analysis of all the DNA in a living cell, as well as gene functioning. In medicine, genomics has the potential to influence medical care and could help individualise treatments. It is currently being used in oncology and pharmacology, and in the treatment of rare and undiagnosed diseases, as well as diagnosed infectious diseases.

Genetic engineering provides the opportunity to produce valuable products of medical importance through recombinant DNA technology. Pharmaceutical products can be developed artificially and more efficiently than with traditional methods of production. For example, *Humulin* (human insulin) is produced for people with diabetes to regulate insulin levels (Fig. 9.18). Before it was developed (in 1982), diabetics were treated with insulin extracted from pigs and cows. The recombinant form of insulin is better tolerated by diabetics and can be produced quickly and efficiently.

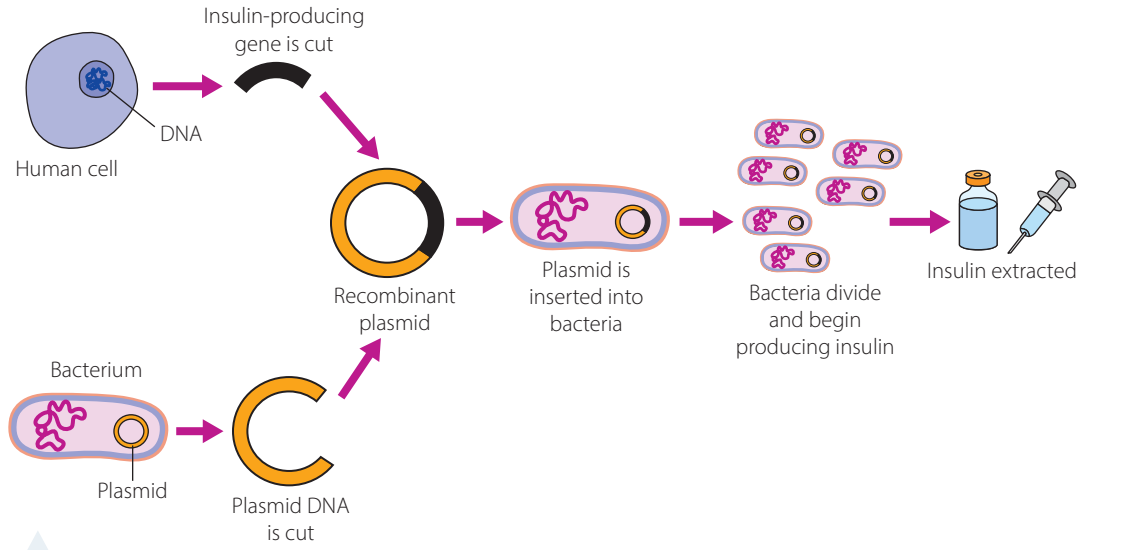

FIGURE 9.18 Recombinant DNA technology used to produce insulin

The role of antibodies in recognising specific protein markers is discussed in more detail in Chapter 12. Cancer is discussed in more detail in Chapter 15.

Monoclonal antibodies (MABs) are used in medicine, particularly cancer research. MABs incorporate the use of genetic technology to artificially clone antibody-producing cells that target specific antigens. MABs are used in cancer treatment to assist the natural immune system to produce specific antibodies that target cancer cells (Fig. 9.19). MABs are grown either in vitro or in the stomach lining of mice. Their role is to recognise specific types of protein markers on cancer cells. Some MABs are produced to activate the immune system, some block the immune system, some stop cancer cells from dividing and others assist in the delivery of drugs and radiation therapy. They are produced for specific forms of cancer and are therefore antigen specific. Cancer patients can be given MABs intravenously in combination with other therapies.

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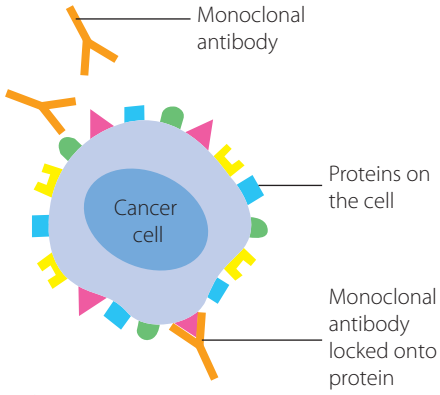
Industrial benefits

Current research projects are aimed at exploring the potential of plants to make compounds for industrial uses. For example, GM plants could be used to produce environmentally friendly chemicals that can replace the non-renewable products that are currently used (such as fuels, plastics and paints). CSIRO has been working on potato plants that produce a type of starch that can be used to produce a range of products such as paper and textiles (Fig. 9.20).

Enzymes, which are biological catalysts, have been used extensively in food production and brewing for at least 6000 years. More recently, recombinant DNA techniques have been used to manufacture enzymes that are used widely in the food industry, particularly in dairy production and brewing. The advantage of using recombinant DNA is that it can be quickly cloned to produce large quantities of an enzyme that is targeted specifically at a particular substrate – an enzyme produced in this way is purer than enzymes produced by more traditional methods, which can contain a range of additional chemical substances.

Researchers have been working on developing genetically engineered plants and bacteria (*E. coli*) that can absorb heavy metals, such as mercury from contaminated sites. This could aid in the remediation of mine sites and other heavily polluted areas, reducing harm to local ecosystems.



Source: Cancer Research UK

FIGURE 9.19 Action of a monoclonal antibody on a cancer cell

FIGURE 9.20 Microscopic image of the surface of a potato plant genetically modified to produce starch for paper, textile and adhesive production

INVESTIGATION 9.4

Benefits and limitations of genetic technologies: A class debate

When evaluating the potential benefits of genetic technologies, you will make a judgement based on certain criteria. The criteria must include reasons for and against. In this investigation you will need to access a variety of sources, including current information from the Internet, scientific journals, videos and other sources.

Ethical understanding

Sustainability

AIMS

1. To gather and process information on examples of genetic technologies in agriculture, medicine and industrial uses
2. To assess the benefits of these technologies and identify some arguments against the use of these biotechnologies
3. To summarise your research in a table
4. To debate some of the benefits and limitations of researching genetic technologies

»

» METHOD

1 Use secondary sources to research in detail at least two examples of genetic technologies in use in agriculture, medicine or industry – for each, make notes on:
   - the details of the two species whose genes have been recombined
   - the advantages and/or disadvantages of this transformation
   - applications in agriculture, medicine and industry
   - the benefits and limitations of the technology.

2 Remember to use sources that are accurate, reliable and valid. Refer to the CRAAP test (pages 10–11). Keep a record of your sources in an appropriate format. Your school library website may be able to provide a suitable template for references gathered. (Acknowledgments and references are also discussed in Chapter 1, page 27.)

3 Prepare to debate the benefits and limitations of genetic technologies. You should be able to argue on the affirmative or the negative team – you will be assigned to a team at the start of the lesson and given a specific topic for debate. Identify *three* advantages and limitations and prepare a written discussion on each.

   (Note: Points for and against *must* link back to the issue, otherwise they will be considered irrelevant.) The table below may be used as a starting point.

	FOR	AGAINST
Agricultural example:		
Medical example:		
Industrial example:		

KEY CONCEPTS

- Evaluating an issue requires you to make a judgement based on certain criteria.
- Potential benefits of using genetic technologies in agriculture include producing crops that are drought resistant and/or pest resistant and produce a higher yield.
- Medical benefits include producing food that has better nutritional value (e.g. golden rice) and the production of pharmaceuticals and vaccines.
- Industrial applications include the potential to produce new polymers, new energy sources and products that are more environmentally friendly.
- Limitations of genetic technologies include their impact on human health and the environment.

CHECK YOUR UNDERSTANDING 9.5

1 Name a technology that induces genetic change, and list two advantages of that technology.
2 Use an example to explain the potential benefit of using genetically modified organisms in the production of food.
3 Use an example to describe the use of a genetically modified organism in medicine.
4 Identify some limitations of using genetic technologies.
5 Discuss the use of genetically modified organisms in industry.

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9.6 The effect of biotechnology in agriculture

Sustainability

Biotechnologies may increase or decrease the genetic diversity of species, depending on how they are used. For example, selective breeding has the potential to both increase and decrease genetic diversity. In the short term, introduced genes broaden the gene pool in a population. In the long term, if selected desirable genes constantly replace other varieties of genes, the gene pool, and therefore genetic diversity, will decrease.

Charles Darwin was aware of the effects of selective breeding – he bred pigeons and used the results of his artificial selection to demonstrate the principles of natural selection. Darwin explained that the process of selection (natural or artificial) determines the success of an individual in reproducing and passing on its variations. In genetic terms, selection acts on the *phenotype* and determines which *genotypes* are passed on, directly affecting the gene pool.

Selective breeding often leads to a reduction in genetic variation – when 'pure-bred' species and animals that are highly 'pedigreed' have been selectively bred for certain 'desirable' traits.

INVESTIGATION 9.5

Secondary-source investigation: Effect of biotechnology on biodiversity

AIM

To evaluate the effect on biodiversity of using biotechnology in agriculture

BACKGROUND

The provision of nutritional food in large quantities is essential to sustain the food demands of the growing global population. GM plant species are often used, with the aim of providing faster-growing and nutritionally superior foods than natural plant species. GM soybeans are generally produced to be resistant to the herbicide glyphosate, the chemical constituent of the herbicide RoundUp. This enables farmers to spray for weeds in the soy plantations without harming the plants. Soybeans are a good source of protein, minerals and fatty acids, and are used in the manufacture of a range of food products including milk, flour, protein and tofu. They are also used in animal feed, and in the manufacture of particle board, adhesives, oils, waxes, lubricants and foam. Soybeans are an important legume worldwide, and an economically important crop in Brazil, the USA and Argentina.

Worksheet
Comparing plant modification methods

GM soy was originally produced as 'RoundUp Ready' soy by the chemical company Monsanto in 1994. It was made using recombinant DNA techniques. In developing the GM soy plant, researchers extracted the desired genes from the bacterium *Agrobacterium tumefaciens*. The genes are inserted into a bacterial plasmid and a gene gun is used to insert the plasmids into the nuclei of soy plant cells. Other companies are now producing similar products using recombinant technologies. The recombinant technique used is similar to that used to produce Bt cotton.

In Brazil, large areas of rainforest have been cleared for soy bean plantations. There is also concern over the effect of glyphosate on the sensitive rainforest ecosystem, due to the extensive use of glyphosate as a herbicide. Glyphosate is safe for human consumption in small amounts and it would be expected that GM soy would include

FIGURE 9.21 Genetically modified soybeans

traces of the herbicide due to spraying of glyphosate on GM soy plantations. Studies in the USA have shown that residual amounts of glyphosate in GM soy products can be quite high, depending on the farming practices, and this is now being monitored.

Some weeds are resistant to glyphosate, and plantations affected by resistant weed growth may require spraying with an additional herbicide, adding to the residual amount of herbicide in the soy product and in the environment. The potential for GM seed dispersal beyond the boundaries of a plantation is possible and is tightly controlled, to prevent resistant strains from affecting ecologically sensitive areas. Run-off of glyphosate into neighbouring rainforest and aquatic environments is possible and could affect sensitive organisms even at low concentrations.

METHOD

Either use the information on GM soy above or select another example of a GM food used in agriculture, to evaluate the effect on biodiversity. In considering biodiversity, consider the biodiversity of the crop as well as the surrounding natural ecosystems. Also consider both long-term and short-term effects.

1 Use reliable sources (Chapter 1, page 10) for this research activity and justify the reliability of each source in an annotated bibliography. A template such as the one below may be used to collect information from each source.

Biotechnology in agriculture: annotated bibliography

Reference:
Why is the source relevant?
What makes this source reliable?
Short summary of the relevant information (no more than two paragraphs):

2 Use secondary sources to research in detail at least two examples of genetic technologies used in agriculture.

Remember to use sources that provide information that is accurate, reliable and valid (Chapter 1, pages 16–17).

RESULTS

1 Summarise your research in a table of arguments for and against the use of your selected GM food in agriculture, based on its impact on biodiversity.

2 Choose a suitable method for representing the information you have collected.

For example, a mind map might be an effective way of completing your evaluation. To make a mind map, see the weblink.

Weblink
Mind map

DISCUSSION

What impact has your selected GM food had on biodiversity in both the short term and the long term?

KEY CONCEPTS

- When reviewing the impact of genetic techniques on biodiversity, it is important to consider both long-term and short-term effects.
- In the short term, introducing a new genotype will increase the biodiversity in a population.
- In the long term, introducing a new genotype may reduce the biodiversity in a population, as large numbers of identical organisms with particular (beneficial) characteristics are created.

CHECK YOUR UNDERSTANDING 9.6

1 What is the long-term impact of using a genetic biotechnology on the biodiversity of a population?
2 Outline the possible effect of artificial pollination on genetic diversity.
3 How can the process of genetic modification affect genetic diversity in a population?
4 Describe a genetic technique used in agriculture and how it affects genetic diversity.

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9.7 Social, economic and cultural influences on biotechnologies

For the past 10 000 years, biotechnologies have been developed to assist humans. Current biotechnologies are complex in their application and have the potential to change society and the environment. In principle, biotechnologies are developed to improve human life. When people make decisions about the influence of a specific biotechnology, they consider their needs, values and priorities. This means that your point of view may differ from someone else's, based on these considerations.

Social context

Social context is the physical and social setting in which we live. In relation to biotechnology, the biotechnological techniques that are available to a society are dictated by the specific needs of that society, as well as the choices made by government, and the wealth of individuals and the economic status of the country. For example, in many countries, DNA fingerprinting techniques are used extensively in forensic science and in paternity testing. In these techniques, non-coding segments of DNA are used to determine differences and similarities. The most common technique is the use of **short tandem repeat (STR)** analysis of DNA.

Short tandem repeats (STR) are a type of sequence marker on a chromosome. Repeats are often 2–10 bases long and vary in number in the population. Primers are used to locate these repeats and are designed to match areas of interest on the chromosome. Primers include a short segment of complementary bases that target the complementary sequence of DNA. The STRs in alleles are counted, to determine the number of shared alleles between samples (Fig. 9.22).

DNA analysis techniques such as STR enable investigators to match samples and solve problems with a high degree of accuracy. However, the process is time consuming and costly, and may not be available in all parts of the world. In addition, ethical issues arising from the use of this technology include the potential for discrimination, as well as ownership and privacy issues in relation to the genetic information obtained, and the possibility of inappropriate applications of the technology.

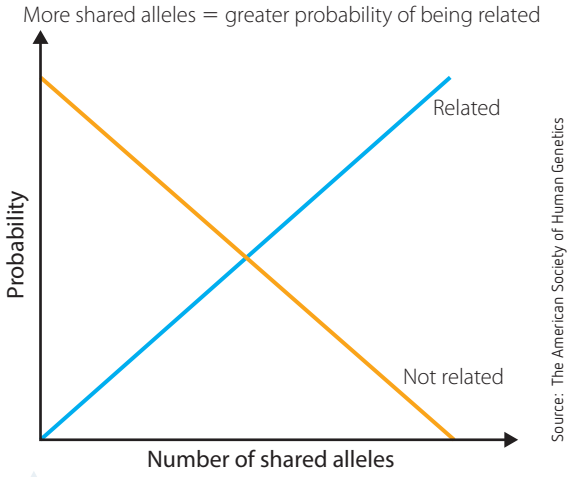

FIGURE 9.22 Determining relatedness between DNA samples: the probability of finding many shared alleles is high in related individuals and low in unrelated individuals.

Economic context

The amount of research and development required to bring genetically modified foods to the consumer is huge. While farmers want to make a profit, there is also a need for consumers to buy food at reasonable prices. Genetically modified products are often patented, which means that the biotechnology company owns the rights to a particular technology. Some argue that this gives large multinational corporations a monopoly. There are growing concerns, for example, that small-scale farmers in developing countries cannot afford to buy the seeds for genetically modified crops, and that this perpetuates the unequal distribution of wealth between developed and developing nations. In some cases, however, GM food can be produced in greater volumes for the same or less cost, so the farmer receives greater financial returns and the consumer pays less. For example, the potential to produce GM Atlantic salmon in greater volumes than are possible in the natural species means the cost of salmon can be reduced for the consumer, and the income of Atlantic salmon farmers can be increased.

FIGURE 9.23 Fossil evidence: 20 000-year-old human footprint at Mungo National Park

Cultural context

Cultural context is dependent on the shared meanings, ideas, beliefs and characteristics of the people who make up a society. The values, religious and moral beliefs and educational background of a group of people may influence their opinions and whether they are open to using biotechnologies.

DNA testing can be used to determine the lineage of fossilised human remains. The results of mitochondrial DNA testing of the fossil remains of Mungo Man, found at Lake Mungo in NSW were first published in 2001 and raised the possibility that another lineage of modern humans inhabited the continent before Aboriginal people. The reliability of the study was recently disputed as the samples were thought to have been contaminated in the original study. In 2016, DNA testing of the fossilised human remains revealed that Mungo Man was in fact of Aboriginal lineage and proved that Aboriginal peoples were the first inhabitants of the Australian continent. The 2016 study used improved DNA testing techniques and compared the DNA of the samples with that of the current inhabitants of the location. The DNA testing and permission to retest the remains of Mungo Man were agreed to by the traditional custodians of Mungo Man, the Muthi Muthi people.

Aboriginal and Torres Strait Islander histories and cultures

INVESTIGATION 9.6

Using secondary sources to assess social, economic and cultural influences on the use of biotechnologies

Ethical understanding

Information and communication technology capability

Advances in biotechnology in areas such as stem cell research and cloning have created high expectations about the possibility of curing diseases such as cancer and repairing worn and damaged tissues, possibly saving millions of human lives. However, not everyone is in favour of all new techniques. For example, for religious or ethical reasons, some people are opposed to cloning that destroys embryos, or the termination of pregnancies where the embryo has been diagnosed with a genetic abnormality. There are strong arguments about the sanctity of human life and humans meddling with nature. In some countries, religion is strongly tied to public policy and public opinion, and has a powerful influence on the regulation of biotechnologies.

In 2010, a survey of people in 32 European countries reflected a strong relationship between strong belief in God and strong opposition to synthetic biology, due to the perception that the creation of a new type of organism was in direct conflict with the idea that creation is a divine activity.

Different religions hold slightly differing views on the development of biotechnology. For example, Islam forbids the consumption of pork, and therefore scholars have raised concerns regarding the introduction of pig genes into food sources or in the development of organs for transplants.

The use of biotechnologies can be an area of contention and controversy, and for this reason you will need to consider the social and cultural factors that influence or bias people's perspectives. For this investigation, you will research the use of one biotechnology.

AIMS

Intercultural understanding

Difference and diversity

1. To use a range of resources in gathering information to assess the social, economic and cultural impacts of the use of one biotechnology
2. To assess the accuracy, validity and reliability of the information gathered
3. To peer review another student's investigation

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METHOD

1. Choose a biotechnology to investigate. Some examples that you may want to consider include:
   - xenotransplantation
   - using salmon genes to create a recombinant DNA strawberry
   - the potential for human cloning
   - IVF.
2. Use a range of resources including books, journals, videos and Internet sources to gather information about the social, economic and cultural impacts on your chosen biotechnology:
   - social impacts – include human rights, benefits to humans, sustainability of resources
   - economic impacts – include financial costs, financial benefits
   - cultural impacts – include religious opinion, difference of opinion due to location and customs.
3. Use a table like the one below to summarise your findings.

	IMPACTS OF BIOTECHNOLOGY
SOCIAL	
ECONOMIC	
CULTURAL	

4. Develop a reference list. (Chapter 1, page 27.)
5. Assess the validity, reliability and accuracy of the sources you used. Use the following as a guide.

ACCURACY	Is the information substantiated? This means is it consistent (reliable) and valid, across more than one source, including scientific literature?
RELIABILITY	Is the information current? Is the source reputable? Check the URL. Does the article aim to create bias? (Look at the author's credentials, date of publication and the organisation(s) they are affiliated with.)
VALIDITY	Does the information relate to the problem you are researching?

6. Working with another student, peer review each other's investigation using agreed criteria (page 4).

CONCLUSION

Use the information you have gathered to assess the overall cultural, economic and social impacts of your chosen biotechnology.

KEY CONCEPTS

- Social, economic and cultural contexts influence the development of biotechnologies.
- These influences vary depending on the biotechnology and the country in which it is developed.

CHECK YOUR UNDERSTANDING 9.7

1. Outline a social influence on the development of biotechnology.
2. Describe an economic influence on the development of a specific named biotechnology.
3. How do different religions view the development of genetic biotechnology?

9 CHAPTER SUMMARY

Genetic technologies: Does artificial manipulation of DNA have the potential to change populations forever?

GENETIC TECHNOLOGIES: ARTIFICIAL MANIPULATION OF DNA

Selective breeding

Choosing desirable characteristics (e.g. crossing Friesian and Jersey cattle)

Artificial insemination

Collecting sperm from a selected male and inseminating several selected females

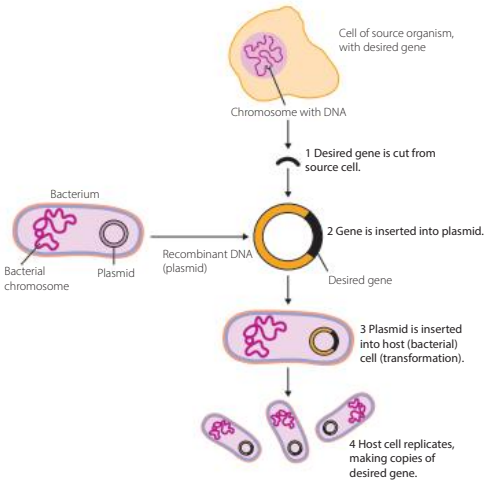
Artificial pollination

Selective breeding in plants by controlling pollination

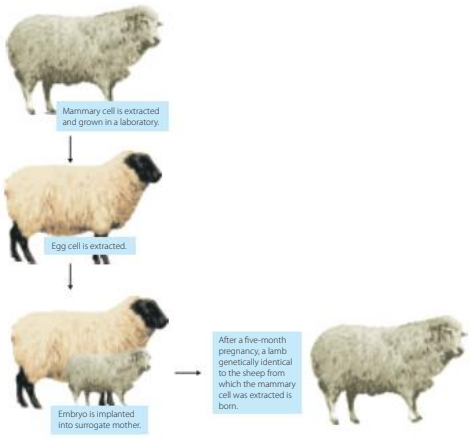
Cloning

Production of an exact copy

Gene cloning – production of identical copies of one gene (e.g. to produce insulin)



Whole organism cloning – cloning adult cells (e.g. somatic nuclear transfer, which involves three animals)



ETHICAL ISSUES AND CLONING

Animal welfare

Concern for the treatment of animals

Moral and religious issues

Same techniques could be used to clone humans

Legal concerns

Who owns the patents to create living things?

Alters genetic composition

Decreases genetic diversity in a population

Cost and access

To the technology

Health risks

Still to be determined

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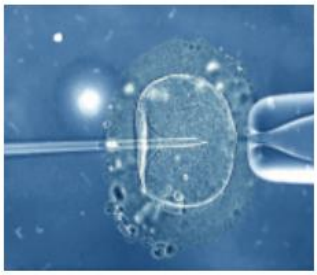
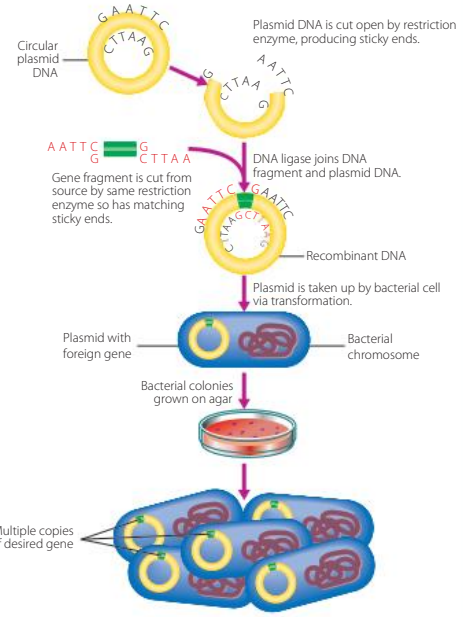
RECOMBINANT DNA TECHNOLOGIES

Methods used to insert the desired gene:

- microinjection
- biolistics
- electroporation
- transduction



Involve inserting the gene of one organism into the genome of another

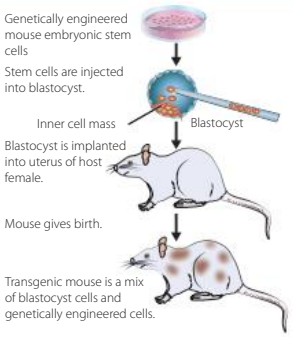


Transgenic species

Created by moving a gene from one species into another (e.g. Bt Cotton)

Medical uses

- Production of vaccines
  e.g. hepatitis B vaccine
- Transgenic sheep produce blood-clotting factor
- Knock-out mice used to study diseases such as Alzheimer's



Agricultural

- Enhances nutrient levels in plants, e.g. golden rice and GM soybean
- Potential to produce GM animals as food, e.g. salmon

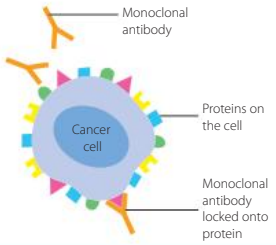
Industrial

- Potential to produce environmentally friendly chemicals
- Replace non-renewable resources

BENEFITS OF USING GENETIC TECHNOLOGIES

Medical

- Opportunity to produce pharmaceuticals, e.g. insulin for diabetics
- Production of monoclonal antibodies for use in cancer treatment



Economic context

- GMO are often patented, making them more expensive.
- There can be unequal distribution of wealth.

EFFECTS OF GENETIC TECHNOLOGIES

Cultural context

Values and beliefs influences opinions about biotechnologies.

Impact on genetic diversity Can increase genetic diversity in the short term but replace varieties of genes in the long term, therefore reducing biodiversity.

Social context Specific needs of a society determine which biotechnologies are used.

9 CHAPTER REVIEW QUESTIONS

Review quiz

1. Outline some examples of current genetic technologies that can induce genetic change.
2. How is artificial insemination used in the conservation of endangered species?
3. Distinguish between artificial pollination and artificial insemination.
4. Explain how the genetic composition of a population can be affected by in vitro fertilisation.
5. How is whole-organism cloning different from gene cloning?
6. Use an example to outline the process of gene cloning.
7. Describe one technique used in the application of recombinant DNA.
8. Identify three applications of transgenic organisms.
9. Draw a flow chart to describe how a transgenic organism can be produced.
10. Explain how transgenic organisms can be useful in a medical application.
11. How is biotechnology used to monitor biodiversity in a population?
12. Why did scientists ensure that the nucleus was removed from an unfertilised egg that was used when making Dolly the sheep?
13. Assess the benefits of using a named biotechnology in the production of food for humans.
14. Why are some people opposed to the use of genetically modified organisms in the production of food or medicine?
15. Explain how biodiversity can potentially be affected by using techniques such as artificial selection and cloning.
16. Genetic manipulation is used to create transgenic species in agriculture. Discuss the ethical considerations of applying this technology to improve a named agricultural product.
17. Explain how the genetic diversity of a species may be increased using:
    a cloning **b** transgenic species.
18. Does artificial manipulation of DNA have the potential to change populations forever? Explain.

Exam preparation

» END-OF-MODULE REVIEW

MODULE 6: GENETIC CHANGE AND BIODIVERSITY

Answer the following questions.

1. Human insulin is made in a recombinant DNA process. The processes involved are listed in A to E below. What is the correct sequence of these processes to make insulin? Write the letters in the order of the steps.
   A The donor DNA fragment from the pancreas is pasted into the plasmid by DNA ligase.
   B A piece of DNA is cut to remove the insulin-producing section of the gene from a human pancreas cell.
   C The bacterial plasmid produces human insulin as the gene spliced into it is expressed. The insulin is harvested for use in human diabetics.
   D The plasmid with its new gene is returned to its bacterial cell, where it multiplies by binary fission.
   E A bacterial plasmid is removed from its cell and cut with a restriction enzyme.
2. Discuss the impact of mutations in DNA repair genes on the introduction of new alleles in a population.
3. In Chile in 2003, the mummified skeleton of a six to eight year-old female was found, but the skeleton was only 15 cm long. Scientists named the skeleton Ata.
   Identify and outline the biotechnologies that could have been used to enable scientists to arrive at each of the conclusions **a** to **d** below.
   a Ata's genome was compared to those of humans, chimpanzees and Rhesus macaques. The skeleton was confirmed as being human.
   b X and Y chromosome analysis revealed that the specimen was female.
   c A comparison of the single nucleotide polymorphisms (SNPs) in Ata's genome with those used as markers for distinct geographical populations suggested that her ancestry was Chilean.
   d Ata was most likely a pre-term birth, with premature bone development. DNA mutations linked to dwarfism were found in several genes associated with bone formation and musculoskeletal development.

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4 It was discovered that in African populations living very close to the equator, there is a higher frequency in the salt-saving version of the 3A5 gene. The mutant allele of the 3A5 gene makes a protein that increases the efficiency of an enzyme that breaks down cortisol, a hormone that raises salt levels in the kidneys and helps the body retain water. Give a reason why this may be an advantage for people living in those geographical regions and explain how this could have led to an increase in the frequency of this allele.

5 For the enzymes in the table compare their functions in replication with their application in gene technologies. Name one gene technology in which each is used.

ENZYME	FUNCTION IN REPLICATION	APPLICATION IN GENE TECHNOLOGY	NAME OF GENE TECHNOLOGY
DNA polymerase			
Ligase			

6 Some biotechnologies have been developed to manipulate DNA, whereas other biotechnologies have been developed to analyse and visualise DNA. Explain the difference between these processes and give an example of a gene technology for each.

7 Explain the application and implications of each of the following uses of biotechnology for conservation and sustainability of biological diversity. You may use examples to help your explanation.

- **a** banking germ plasm from plants
- **b** use of genetically modified crops as food
- **c** mapping relatedness of animals in the wild and in captivity for breeding programs
- **d** artificial insemination for conservation in zoos and in the wild

8 Explain the importance of understanding endocrinology (how hormones function) and the process of IVF to conduct gene therapy as a medical intervention to correct defective genes in offspring.

9 Compare the process of PCR in a laboratory with DNA replication in cells.

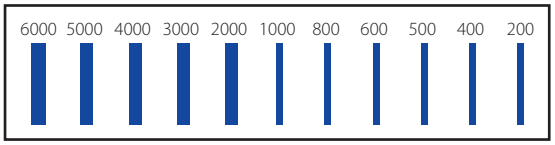
10 Explain how identifying end-product proteins produced by a cell can give insight into the genome of that cell.

11 Explain why, in DNA analysis techniques, a DNA probe needs to be labelled with a molecular tag (such as a radioactive atom or a fluorescent dye).

12 Biofuel can be produced by fermenting sugars from plants such as sugar cane, corn, wheat, canola and sugar beet to ethanol. It uses a similar process to that used in beer and wine-making. If these biofuels are blended with petrol or diesel, they reduce the amount of carbon emissions. Propose reasons for and against furthering this type of research for the production of fuels for aviation.

13 A DNA fragment with a total length of 3000 base pairs is cut using restriction enzymes at the positions shown by the arrows. The numbers in brackets indicate the number of bases in each fragment after cutting.

DNA bands of known size were created by using the restriction enzyme EcoR1 and run through a gel by electrophoresis. Copy the diagram and mark where bands from the sample DNA fragment would align on the gel.



14 The table in the Key concepts box on page 253 (Chapter 7) lists factors such as selective pressure, sexual selection, mutation, genetic drift and gene flow. Using this table, provide an example to illustrate each 'change in allele' described. Use information from Chapters 1–7 to find examples, and keep a record of which chapter you found the information in, for each.

DEPTH STUDY SUGGESTIONS

- → Investigate a specific genetic biotechnology and how it can be used to treat a particular human disease.
- → Research the ethics of xenotransplantation and the laws that regulate it.
- → How can cloning have the potential to bring back extinct organisms?
- → Myth bust: Is Jurassic Park possible, with today's biotechnologies?
- → Create a survey to study a social, cultural or economic influence on biotechnology development.

» MODULE SEVEN

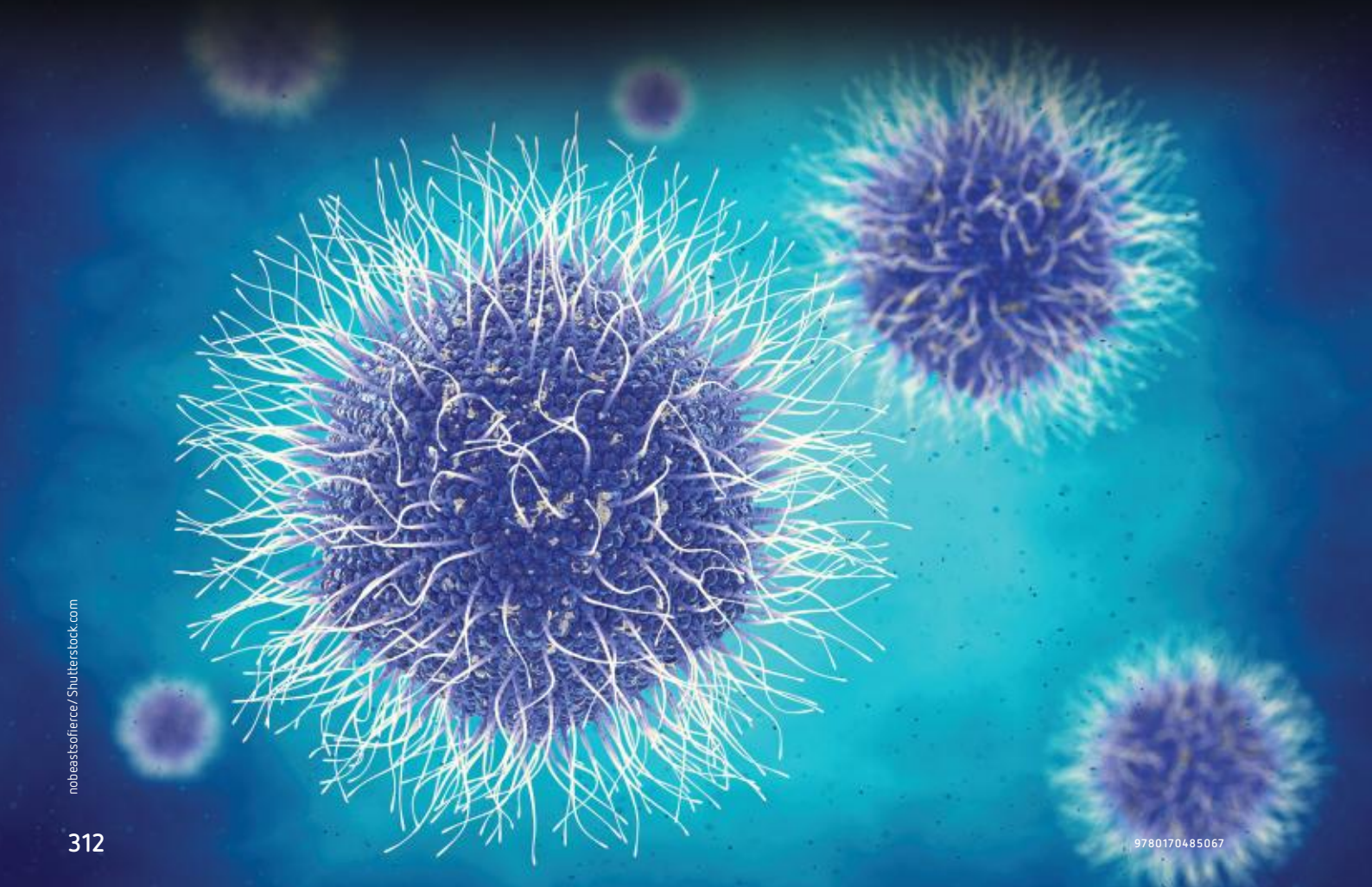
INFECTIOUS DISEASE

10 Cause and transmission of infectious disease

11 Responses to pathogens

12 Immunity

13 Prevention, treatment and control of disease



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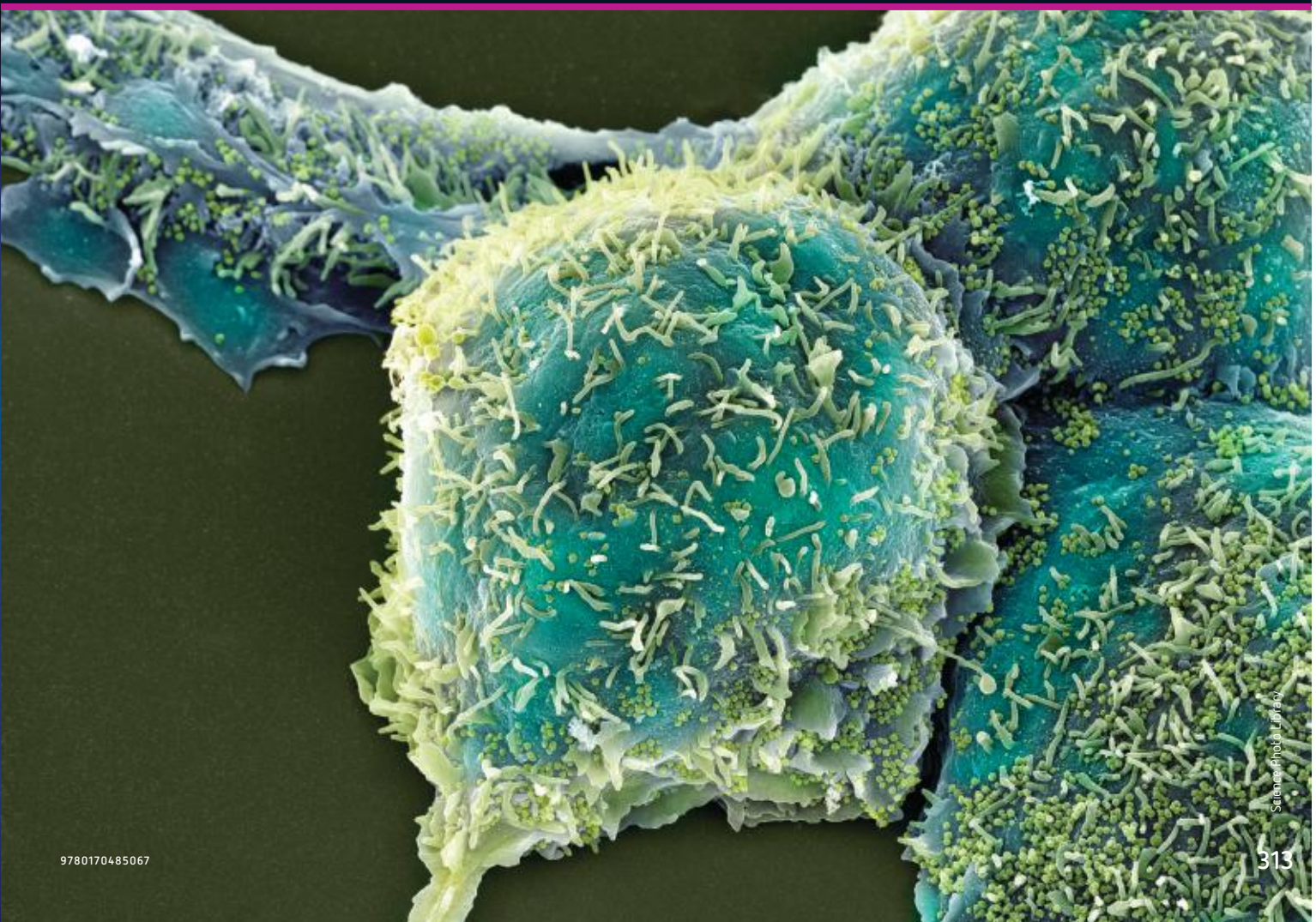
10 Cause and transmission of infectious disease

INQUIRY QUESTION

How are diseases transmitted?

Students:

- describe a variety of infectious diseases caused by pathogens, including micro-organisms, macroorganisms and non-cellular pathogens, and collect primary and secondary-sourced data and information relating to disease transmission, including: (ACSBL098, ACSBL097, ACSBL116, ACSBL117)
  - classifying different pathogens that cause disease in plants and animals (ACSBL117)
  - investigating the transmission of a disease during an epidemic
  - design and conduct a practical investigation relating to the microbial testing of water or food samples ICT
  - investigate modes of transmission of infectious diseases, including direct contact, indirect contact and vector transmission
- investigate of the work of Robert Koch and Louis Pasteur, to explain the causes and transmission of infectious diseases, including: L, WE
  - Koch's postulates
  - Pasteur's experiments on microbial contamination
- assess the causes and effects of diseases on agricultural production, including but not limited to: S, WE
  - plant diseases
  - animal diseases
- compare the adaptations of different pathogens that facilitate their entry into and transmission between hosts (ACSBL118)



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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
10.1 A primary investigation of the comparative sizes of pathogenic cells
10.2 Secondary-source investigation of transmission of a disease during an epidemic
10.3 Primary investigation into the microbial testing of food and water samples
10.4 Primary investigation to model Pasteur's swan-necked flask experiment
10.5 Secondary-source investigation of plant diseases and their transmission
10.6 Secondary-source activity on diseases of agricultural significance in Australia

Worksheets
- Diversity of pathogens
- The importance of sterile technique
- Koch's postulates
- Louis Pasteur and the germ theory of disease
- The risk of infectious disease in agriculture

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To access these resources, visit **cengage.com.au/nelsonmindtap**


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FIGURE 10.1 As in a game of chess, the body relies on many strategies to deal with harmful microbes.

For many years, the interaction between humans and the organisms that seek to inhabit them has been portrayed as a constant and fierce battle between deadly enemies, ending only with the elimination of one side. This metaphor fails to acknowledge the subtlety of the immune response and the ability of an organism's combined defences to survey newly recognised molecules and instigate a measured and effective response. If our immune system declared all-out war every time, the result would be chronic, unresolvable inflammation in all tissues exposed to the outside world, and poor health. Worse still, the immune system in this overactive state could direct its defences against the organism itself, as occurs in **autoimmune diseases** such as lupus and type I diabetes mellitus, or give rise to allergies such as asthma. Instead, the systems that protect multicellular organisms from outside threats are less like a war, and more like a game of chess, where the players plan ahead, and know when to advance and retreat.

10.1 The variety of infectious diseases caused by pathogens

Prior to the work of scientists such as Louis Pasteur and Robert Koch, little was understood about the nature of infectious disease. The 'germ theory of disease', that disease and decay are the product of living organisms, was unknown to people, who still believed in **spontaneous generation** – that living organisms could arise from inanimate material such as rotting meat. As with most scientific discoveries, the mystery of infectious disease was unlocked progressively, as the science of **microbiology** emerged. Critical to the development of this science was the concurrent development of technology such as the light microscope, as well as techniques in staining and in sampling diseased tissue.

Disease is any process or condition that adversely affects the normal functioning of a living thing or parts of a living thing. An **infectious disease** is caused by another organism or an infective **agent** known as a **pathogen** (and is thought of as being 'caught' from somewhere or someone). Pathogens may be single-celled microbes such as bacteria, protozoa and yeasts, multicellular parasites such as fungi and worms,

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or non-cellular agents such as viruses and prions. Measles and influenza are examples of infectious diseases of humans. Infectious diseases are distinguished from **non-infectious diseases** such as cancer and diabetes, which do not involve the transfer of a pathogen between hosts and therefore cannot be 'caught'. There are exceptions to this, however. For example, devil facial tumour disease is a cancer that is also infectious, because cancer cells are transferred when one infected Tasmanian devil bites another.

The term **communicable disease** is often used to describe a disease that can be transmitted from plant to plant, or from animal to animal. Pathogens have a range of strategies to achieve this transfer, based on molecules known as **virulence factors**, which enable the pathogen to inhabit its host more effectively and may even enable it to evade destruction by antibiotics. An example is bacteria that produce the enzyme beta-lactamase and are resistant to penicillin-derived antibiotics.

What is disease?

The word 'disease' derives from the old French terms *des* and *aise*, meaning a 'lack of ease'. Disease processes are typified by a series of signs and **symptoms**. Signs are objectively measurable factors, such as increased body temperature (pyrexia or fever), the appearance of a rash, an increase in blood pressure (hypertension) or a change in the colour of a tissue. When tissue is damaged, it forms a **lesion** (such as a wound or an abscess). Signs are usually recorded during a physical examination of the body systems and include the results of any **histopathology** that may be undertaken. Symptoms are factors reported by the patient, such as pain, fatigue, nausea or headache, that cannot be directly observed by others. A combination of signs and symptoms gives a complete picture of the effects of pathogens on the organism.

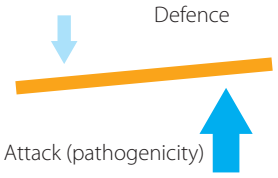


FIGURE 10.2 Disease is the result of a loss of balance between defence and attack.

In low numbers, a pathogen may be incapable of initiating a disease. Increases in pathogenic **burden** are often associated with infectious disease, as the body's immune defences become overwhelmed. The likelihood of an organism developing an infectious disease depends on the balance between:

- the **pathogenicity** of the microbe, including the numbers of pathogens ('burden')
- the defence capabilities of the host (Figure 10.2).

An example is the occurrence of a type of mange in dogs caused by the mite *Demodex canis*. This mite is a normal inhabitant of the hair follicles of canines and is usually present in low numbers. The clinical expression of demodectic mange (Fig. 10.3) is most common in young puppies whose immune systems are still developing, or in older animals whose immune systems are compromised. In these cases, the mite acts as an **opportunistic** pathogen – this can happen if there are concurrent diseases, or if the animal has been given a course of drugs that have a suppressive effect on the immune system, such as corticosteroids, which are commonly prescribed for chronic skin inflammation.

FIGURE 10.3 The mange mite *Demodex canis* becomes pathogenic only when immune suppression occurs.

KEY CONCEPTS

- The immune system is a complex network of responses to outside threats.
- Infectious diseases involve threats from infectious agents known as pathogens.
- There is a fine balance between an appropriate and useful response and one that may harm the host organism.
- Disease results when there is a loss of balance between defence and attack.

Classifying pathogens

Pathogens can be broadly classified as shown in figures 10.4 and 10.5.

FIGURE 10.4 Broad classification of pathogens

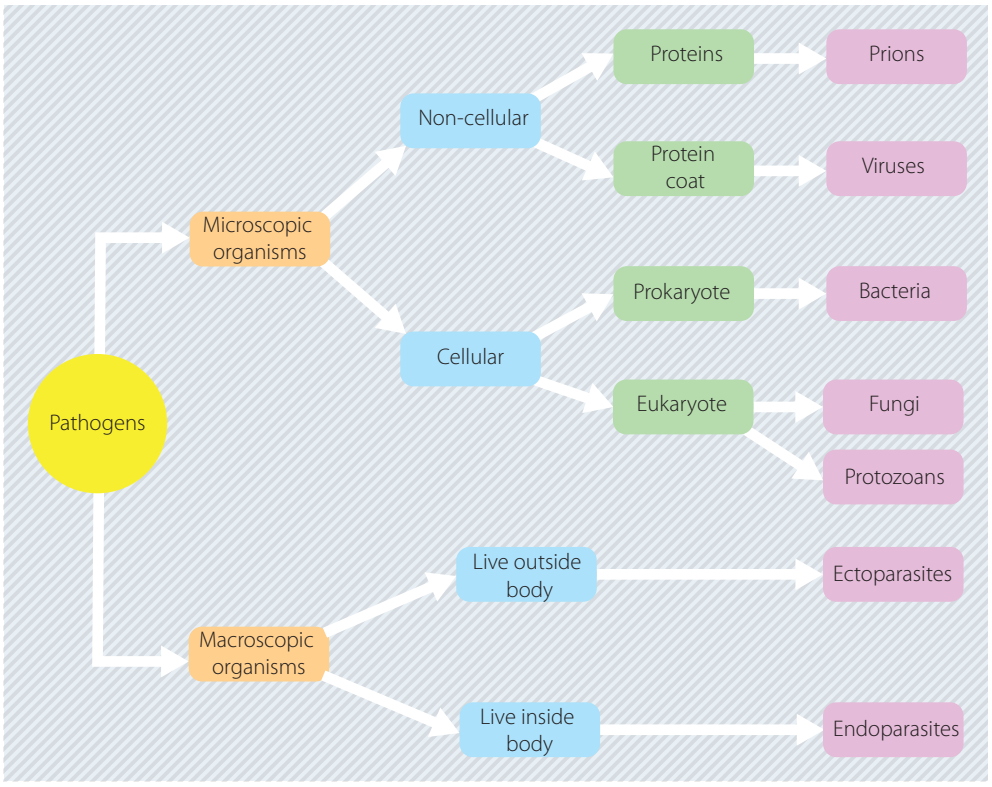
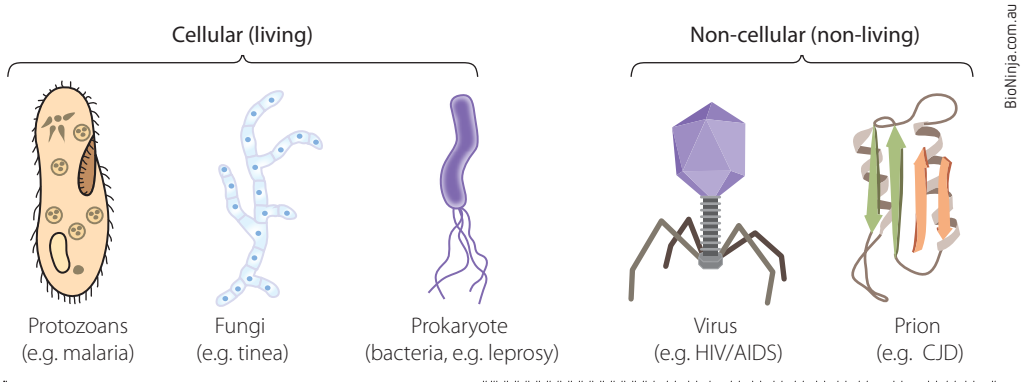


FIGURE 10.5 Cellular and non-cellular pathogens, and examples of diseases they cause (CJD: Creutzfeldt-Jacob disease)



Cellular pathogens: meningococcal meningitis

In 2017, the NSW Government implemented a school-based vaccination program for Year 11 and 12 students, to provide protection against a significant threat to their health. Meningococcal disease is a serious bacterial disease that can cause acute illness and death due to bacterial **meningitis** (bacteria in the brain) and **septicaemia** (bacteria in the blood). A new form of the bacteria, known as type W, had resulted in a threefold increase in notifications of the disease during the school year and was causing concern among health professionals. A decade of vaccination against other strains of the disease had seen a decrease in the frequency of the disease. The most common symptoms of meningococcal disease include fever, headache and stiffness of the neck, as well as pain in the joints. A red-purple rash (Fig. 10.6) is common, as well as nausea and a dislike of bright lights (**photophobia**). Older teenagers and young

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adults are at increased risk of contracting the disease, due to social behaviours that increase the risk of transmission. Rapid treatment is vital, to preserve life and minimise damage to organ systems.

A vaccine is now available that covers meningococcal types A, C, W and Y. The vaccine is particularly recommended for young people living in close quarters, such as military establishments and boarding schools.

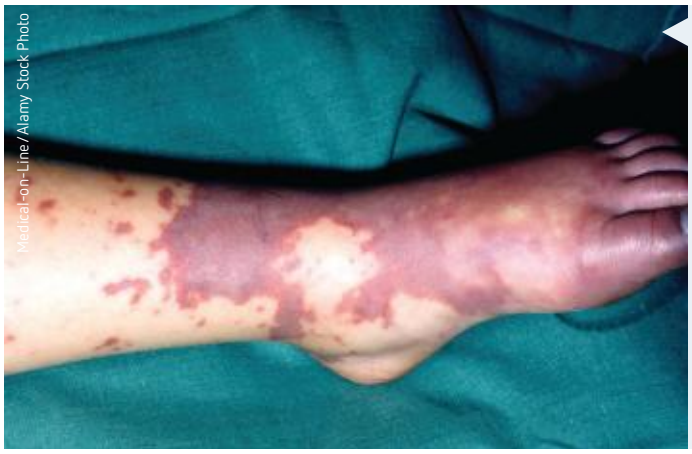

FIGURE 10.6 A severe rash that does not blanch (go pale) when pressed is a sign of meningococcal disease.

Classification of bacteria

Bacteria are single-celled **prokaryotic** organisms. They have a cell wall but no membrane-bound nucleus or organelles (Fig. 10.7). They are classified as one of the three **domains** of living things (eukaryotes, bacteria and archaea).

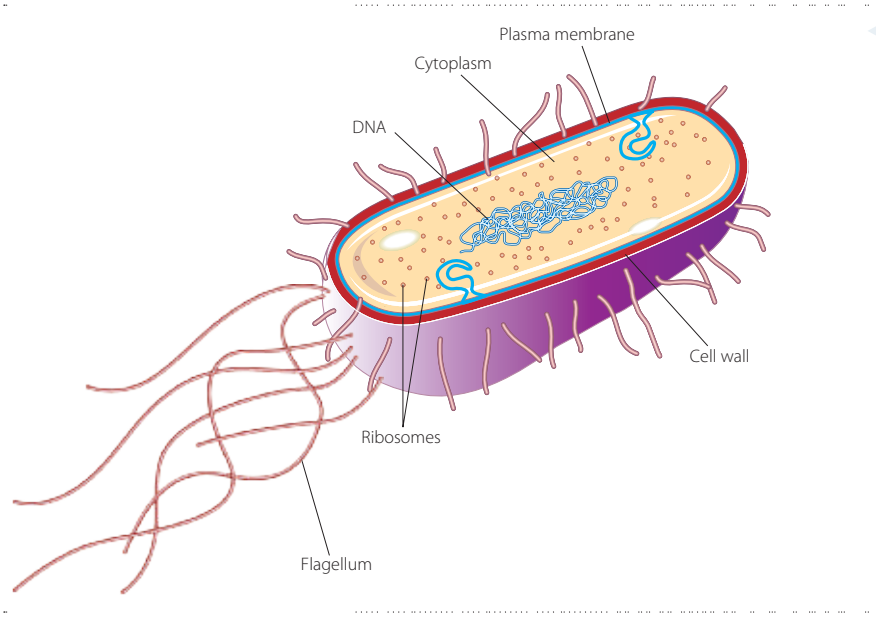

FIGURE 10.7 The general structure of a bacterium

Bacteria reproduce by an asexual process known as binary fission (dividing in two). The time it takes for the number of bacteria to double, known as the *generation time*, varies between species, but is between 10 minutes and 24 hours. This means that huge numbers of bacteria can be produced in a very short time.

Bacteria are larger than viruses but smaller than protozoans, varying in size from 0.2 to 10 µm (micrometres) in length. Their genetic material is in two forms: as bacterial DNA in the form of a large circular chromosome, and as smaller circular DNA fragments called plasmids. Their cell wall is composed of **peptidoglycan**, a substance made of protein and carbohydrate molecules (unlike plant cell walls, which

are composed of cellulose). Some bacteria, such as those in the **genus** *Mycoplasma*, lack a cell wall. The mycoplasmas are **obligate** intracellular parasites – this means they cannot live and reproduce outside a cell. *M. pneumoniae* causes pneumonia in humans, and *M. gallisepticum* causes chronic respiratory disease in poultry and other wild birds. Some bacteria possess a **polysaccharide capsule**, which can act as a virulence factor, making them more effective in causing disease.

Bacteria may also be classified according to their shape (Figs. 10.8, 10.9): spherical (coccus), rod-shaped (bacillus), spiral (spirillum), comma-shaped (vibrio).

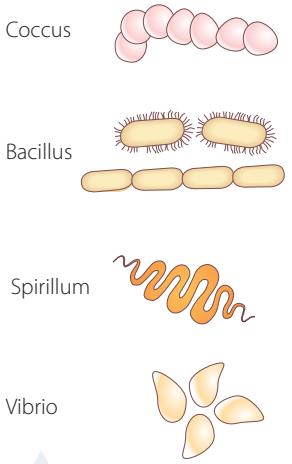

FIGURE 10.8 Different types of bacteria, based on their shape

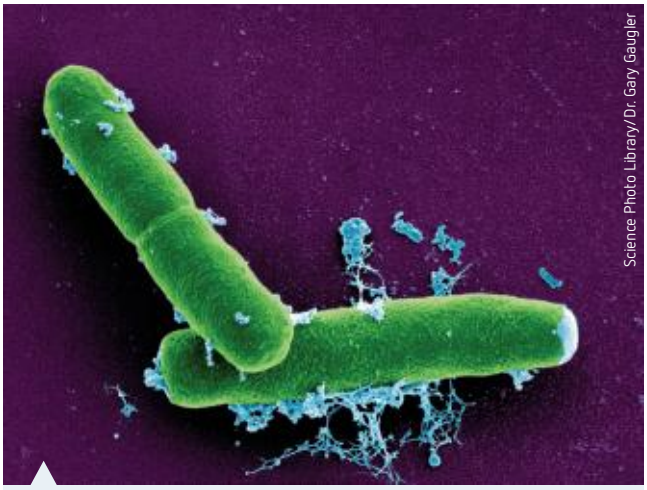

FIGURE 10.9 Example of a rod-shaped bacterium, *Bacillus anthracis*, which causes anthrax

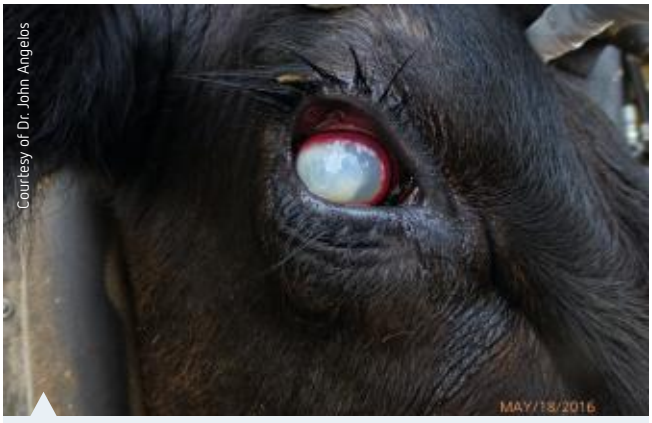

FIGURE 10.10 'Pink eye' in cattle (infectious keratoconjunctivitis) is caused by the Gram-negative bacterium *Moraxella bovis*.

Some bacteria (such as *Rickettsia* and *Chlamydia* species) are obligate intracellular parasites. Others (such as *E.coli* and *Pseudomonas aeruginosa*) are extracellular parasites – they live in tissue fluid.

Different types of bacteria can be distinguished by their staining properties (using Gram stain). Some bacteria are **Gram-positive**, others are **Gram-negative** (Fig. 10.10). Bacteria may be **aerobes** (survive in an oxygenated environment), **anaerobes** or **facultative anaerobes** (survive by aerobic respiration if oxygen is present, but can switch to anaerobic respiration if oxygen is absent).

Transmission of bacterial disease

Bacteria are everywhere in nature, including water and soil, and serve many vital roles in ecosystems. They are also used by humans to create vaccines and antibiotics. They can inhabit multicellular organisms in a beneficial, symbiotic relationship – there are an estimated 1000 trillion bacteria in the human body and most of these are beneficial.

Transmission of pathogens is discussed in more detail later in the chapter.

Bacteria that cause disease do so by producing toxins or chemicals that are harmful to the host's body, or by damaging host tissue directly. This represents a **parasitic** relationship, as there is benefit to the bacteria and harm to the host.

Transmission of bacterial diseases may occur directly through close contact with another infected host organism, or indirectly through contact with an object contaminated with the bacterium.

Many bacteria are susceptible to chemical treatment with antibiotics, with concurrent management such as surgical removal of dead tissue, wound cleansing and other supportive treatment.

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Some bacteria are capable of forming an **endospore** (a tough, waterproof external layer) and lying dormant in the environment for years. **Clostridial bacteria** such as *Clostridium tetani*, the causal agent of tetanus, can lie dormant in soil for decades, especially around horse properties. Endospores can resist heat, chemicals and **desiccation** (drying out), which makes them extremely problematic, as normal methods of sterilisation may be ineffective in eliminating them.

Bacteria may aggregate, forming a mucus-like structure called a **biofilm**. A biofilm can bind to living tissue, and the carbohydrate matrix formed within it can also bind the bacteria to other surfaces, such as implants and catheters. This strategy enhances their defences against antibiotics and can make their detection and elimination very difficult.

TABLE 10.1 Examples of diseases caused by bacterial pathogens

BACTERIA	NAME OF DISEASE	FEATURES OF DISEASE
Bordetella pertussis	Whooping cough (pertussis)	• Runny nose, sneezing • Characteristic 'whoop' during coughing bouts • Gagging or vomiting
Salmonella enterica	Salmonellosis (food poisoning)	• Vomiting and diarrhoea • Dehydration • Fever and abdominal cramps
Mycobacterium tuberculosis	Tuberculosis	• Cough • Fever • Night sweats • Blood-stained sputum
Clostridium tetani	Tetanus ('lockjaw')	• Fever, sweating, headache • Dysphagia (difficulty swallowing) • Tachycardia (rapid heartbeat) • Muscle spasms that begin with jaw and spread to rest of body
Chlamydia trachomatis	Chlamydial disease (sexually transmitted)	• Burning sensation while urinating • Discharge • May lead to infertility if untreated

Fungi: Tinea (athlete's foot)

You may have been advised never to shower at your gym or local pool without wearing 'shower shoes' such as sandals or thongs. The warm and moist environment of a public shower provides ideal conditions for the growth and spread of fungal pathogens. Tinea (*Tinea pedis*) or 'athlete's foot' is a common fungal disease in humans. The symptoms are redness and itching of the skin around the toes, scaling of the skin, and nail deformities such as yellowing and hardening (Fig. 10.11). Treatment involves the use of antifungal creams for an extended period, even after the symptoms are gone.

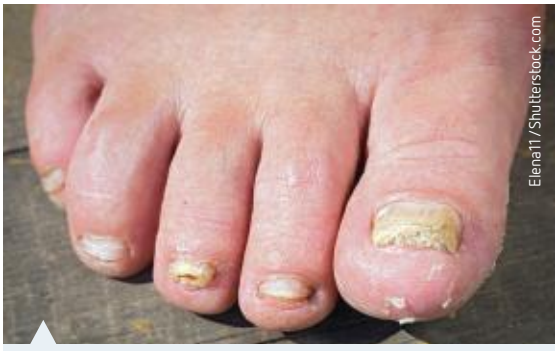

FIGURE 10.11 Tinea can lead to hardening and yellowing of nails.

Classification of fungi

Fungi are eukaryotic organisms that have a cell wall composed of **chitin** (unlike the cellulose cell walls of plants). Fungi are heterotrophic – they do not contain chlorophyll and are not capable of producing their own nutrients. Most types of fungi are **saprophytic** – they live on dead plant and animal material, and therefore play an important role as decomposers in an ecosystem.

Fungi may be unicellular (such as yeasts, Fig. 10.12 and Fig. 2.25a) or multicellular (such as moulds, Fig. 10.13). Moulds are composed of a system of microscopic tubular filaments or threads known as **hyphae**, which branch and spread to form a structure known as a **mycelium** (Fig. 2.31).

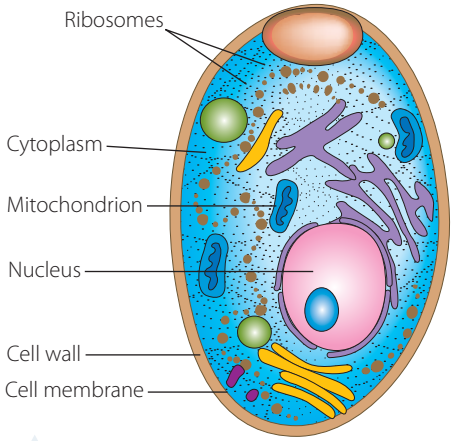

FIGURE 10.12 Structure of a yeast cell as seen under the electron microscope

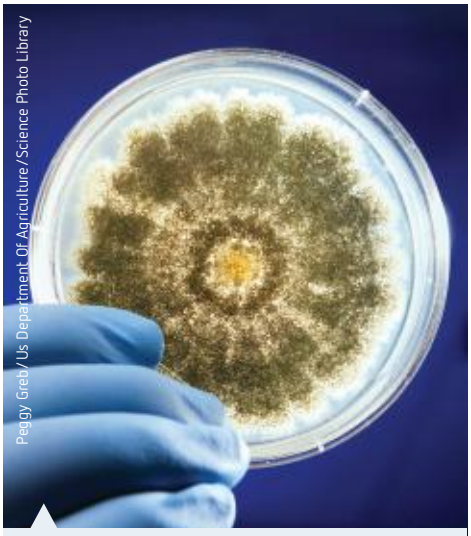

FIGURE 10.13 *Aspergillus flavus*, a multicellular fungus that has a 'fluffy' appearance when grown on an agar plate, caused by the growth of hyphae

Fungi vary in size from microscopic to macroscopic. Some fungi (such as yeasts) reproduce **asexually**, while others reproduce using both sexual and asexual reproduction. Some types (yeasts) are used by humans to produce bread and alcohol.

Transmission of fungal disease

Fungal infections (**mycoses**) may be cutaneous (outer skin layer), subcutaneous (beneath the skin surface, from penetrating wounds) or systemic (affecting internal organs). Most fungal infections are **opportunistic**, affecting those with weakened immune systems or concurrent diseases. Many fungal pathogens are **dermatophytes**, meaning they live on skin, nails and hair. Some fungi, such as *Aspergillus*, *Blastomycosis*, *Cryptococcosis* and *Coccidioides*, can cause serious and life-threatening fungal diseases in humans and other animals. Fungal pathogens may be transferred via close contact with a diseased person or animal, or with contaminated objects. Thrush is a fungal disease caused by a yeast, *Candida albicans* (Fig. 10.14), and can be associated with long-term use of asthma inhalants containing corticosteroids and the long-term use of antibiotics.

In plants, fungi are one of the leading causes of infectious diseases, such as blights, mildews and rusts.

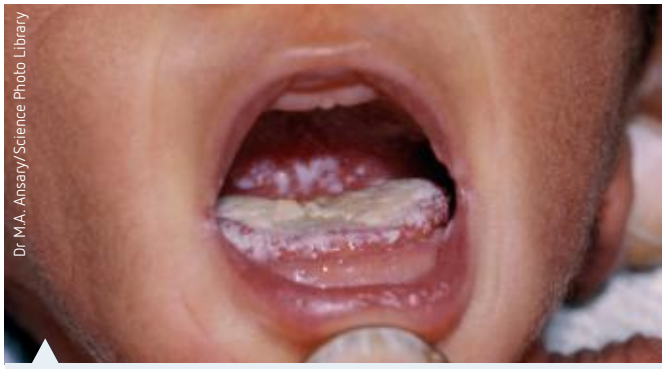

FIGURE 10.14 Oral thrush is a fungal disease caused by a yeast.

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TABLE 10.2 Examples of diseases caused by fungal pathogens (mycoses)

FUNGUS	NAME OF DISEASE	FEATURES OF DISEASE
• *Epidermophyton* • *Trychophyton* • *Microsporum*	• Tinea • Tinea • Ringworm	• Cutaneous mycoses • Fungus secretes enzymes that break down keratin • Exposure to pathogen combined with warm, moist conditions necessary • Redness, itching, scaliness of skin • Ringworm may cause classic ring-shaped lesions on skin • Yellowing and hardening of nails
• *Sporothrix schenckii*	• Sporotrichosis (rose gardener's disease)	• Subcutaneous mycosis • Small cut or scrape allows fungal entry • Scratches from thorns combined with handling infected plant matter • Scratches and bites from cats
• *Histoplasma capsulatum* • *Blastomyces dermatidis* • *Coccidioides* spp.	• Histoplasmosis • Blastomycosis • Coccidiodomycosis	• Systemic mycoses that cause severe lung disease • Histoplasma grows in soil contaminated with bat droppings • Blastomycosis is contracted by breathing in fungal spores from moist soil and leaf material

Protozoa: giardiasis and cyptosporidiosis

Swimming is a favourite pastime of many Australians. In summer, local pools and splash parks are filled with families trying to escape the heat (Fig. 10.15). Unfortunately, when humans are crowded into one place there is an increased risk of the transfer of protozoal pathogens from infected people to others. (Protozoa, a sub-group of unicelluar Protista, are microscopic animal-like unicellular organisms.) *Cryptosporidium parvum* and *Giardia lamblia* (Fig. 10.16) are protozoal organisms that live in the human gastrointestinal tract and are possible sewage and pool contaminants. Infection causes abdominal cramps, chronic diarrhoea, bloating and weight loss. Both organisms are resistant to chlorine under normal pool operating conditions. Infected infants and children are a particular source of the pathogen, due to faecal contamination of pools. Swallowing infected pool water is the main source of transmission.

Boris Sosnovyy/Shutterstock.com

FIGURE 10.15 Splash parks are a potential source of protozoal pathogens if not managed carefully.

During the summer of 2016, an outbreak of cryptosporidiosis prompted a warning from health authorities to councils regarding the maintenance of splash parks and interactive water fountains. They were suspected to be the source of the outbreaks, leading to the media dubbing them 'poo parks'. Plans were made by the NSW State Government to include splash parks under the definition of 'public swimming pools' so that strict water quality controls could be implemented and monitored closely.

Transmission of protozoa

Diseases caused by protozoans include malaria and trypanosomiasis (African sleeping sickness), which are transmitted by insect bites. Amoebic dysentery and giardiasis are transmitted in contaminated water (faeco-oral route).

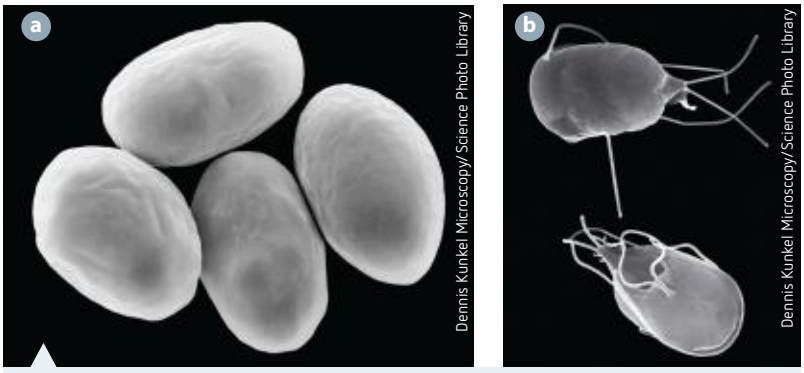

FIGURE 10.16 *Giardia lamblia*, a protozoan and potential pool contaminant: **a** eggs; **b** adults

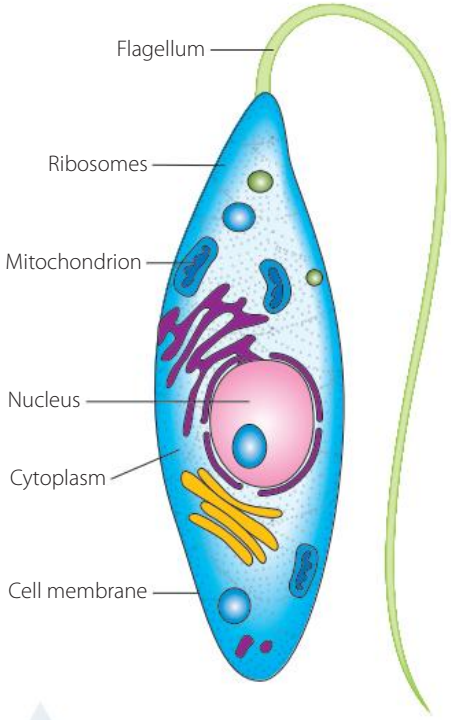

FIGURE 10.17 A generalised protozoal cell

Classification of protozoa

Protozoa are single-celled eukaryotic organisms classified along with algae and slime moulds in the Kingdom Protista (Chapter 2). They have a membrane-bound nucleus, membrane-bound organelles and a cell membrane, but no cell wall (Fig. 10.17). They usually reproduce by the process of binary fission. They range in size from 1 to 300 µm, with the smallest the size of a bacterium. Most protozoans are motile, and their type of locomotion forms the basis of their classification (Fig. 10.18). **Flagellates** are propelled by a long whip-like tail called a flagellum. **Ciliates** have many hair-like projections, called cilia, which propel the protozoan by beating rapidly. **Amoebae** use projections of the cytoplasm, called pseudopods, to move around. **Sporozoa** (such as *Plasmodium*) are protozoans that do not have structures for motion and reproduce by releasing spores.

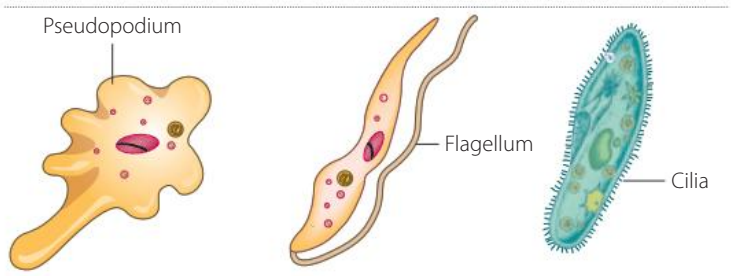

FIGURE 10.18 based on their mode of locomotion

Many protozoa are free-living and do not cause disease, but some are pathogenic (Fig. 10.19, Table 10.3). Protozoal diseases are generally treated using antiprotozoal medications.

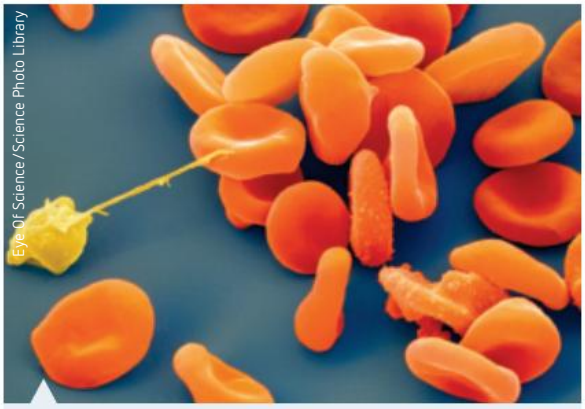

FIGURE 10.19 Red blood cells and *P. falciparum*, one of the species of *Plasmodium* that causes malaria

TABLE 10.3 Examples of diseases caused by protozoal pathogens

PROTOZOAN	NAME OF DISEASE	FEATURES OF DISEASE
Plasmodium spp.	Malaria	• Fever • Fatigue • Headaches • Jaundice • Vomiting
Toxoplasma gondii	Toxoplasmosis	• May be asymptomatic • Headache, fatigue • Fever • Muscle aches • Tender lymph nodes • Seizures
Entamoeba histolytica	Amoebiasis	• May be asymptomatic • Diarrhoea (often bloody) • Fever and chills

Macro-organisms (macroparasites)

Macroparasites are larger than other pathogens – they are visible to the naked eye. They are multicellular eukaryotic organisms, varying in size from the tiniest louse to very long tapeworms. Some macroparasites cause disease directly, while others act as **vectors** in the transmission of disease. They can be classified into two groups, according to where they live.

Endoparasites live inside the host's body. Examples are flatworms (tapeworms and flukes) and roundworms. They cause diseases such as *taeniasis* (tapeworm disease), *hydatidosis* (hydatid disease), *schistosomiasis* (blood fluke disease) and *elephantiasis* (caused by a filarial worm).

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Ectoparasites live on the outside of the body, usually sucking blood. Examples are mosquitoes, fleas, ticks, leeches, mites and lice. Some inject toxins while feeding; these can cause inflammation, allergic reactions and sometimes partial paralysis. Ectoparasites can also act as vectors for the transmission of other pathogens. The flea is a vector for the bacterium *Yersinia pestis*, which causes bubonic plague.

Parasitic helminths: heartworm

In the 1970s, vets in Sydney noticed an increase in the incidence of heartworm in dogs. Heartworms are long, thin worms that live in the right side of the heart as adults and block the flow of blood (Fig. 10.20). They cause damage to the lining of the heart, the valves and the pulmonary veins that exit the right side of the heart. It had been recognised as a problem in tropical north Queensland previously but had not been seen so far south before. Symptoms include a cough, and accumulation of fluid in the abdomen and lungs. The liver was often swollen and affected dogs would suddenly collapse and die. Diagnosis included radiographic detection of classic changes to the lungs and heart, but reliable blood tests have since been developed. Treatment is often as dangerous as the disease, as a mass of dying worms can dislodge from the heart and block major blood vessels. Heartworm is spread by mosquitoes that bite an infected host and transfer it to other dogs. Cats can also be infested with heartworm.

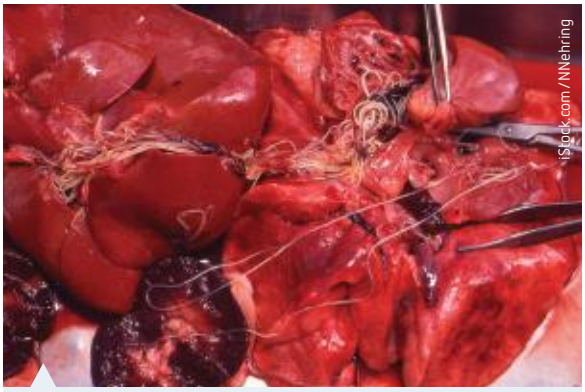

FIGURE 10.20 Adult heartworms in a dog's heart

Classification of helminths

Helminths are worm-like organisms. They commonly inhabit the gastrointestinal systems of humans and other animals, living on nutrients supplied by the host. They disrupt normal digestion and absorption of gut contents. They adversely affect the host organism by the way they attach to the gut wall; the host's immune response is affected, as well as the way it feeds. Worms are generally treated using **anthelmintics**.

The following types of helminths are a significant cause of disease in humans and animals:

- nematodes (roundworms, whipworms, hookworms)
- cestodes (tapeworms)
- flatworms (platyhelminths, such as flukes).

Transmission of helminths

Most helminths have a complex life cycle that involves several stages and several hosts. Part of the life cycle may be spent in the external environment. All helminths reproduce using eggs, which may be deposited in the environment, to be picked up by another host through unsanitary drinking water or infected soil. Larvae hatch from the eggs and mature in the host (Fig. 10.21).

The eggs of each species have a unique architecture, and examination under a microscope can help to identify the species. **Faecal egg counts** are performed on farm animals to estimate the abundance and species of worm parasites in a herd. Helminth infections place an economic burden on agricultural enterprises through animal illness and reduced meat, milk and wool yields in infected livestock. Overcrowding (high stocking rates) is a prime contributing factor to problems with parasitic worms in agricultural enterprises as this facilitates the rapid spread of the parasite eggs. Research suggests that low burdens of parasitic worms may confer an advantage to the host, by stimulating the immune system.

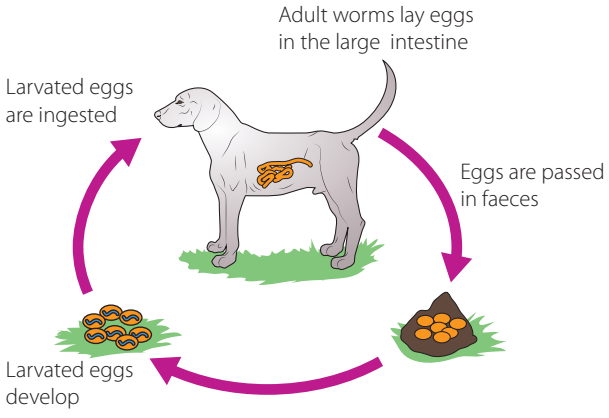

FIGURE 10.21 Intestinal parasites have a long and complex life cycle involving multiple stages. The eggs can survive in the environment for years.

Domestic pets such as cats and dogs are routinely administered anthelmintic drugs, commonly known as 'deworming'. Large burdens of helminths such as hookworm can have dire consequences for puppies and kittens as they cause intestinal bleeding and anaemia, while roundworms can contribute to intestinal blockages and torsions. Sometimes worms will irritate an animal's bottom and lead to the behaviour known as 'scooting' as the animal drags its itchy bottom along the ground. Many worms (such as strongyles or large redworms) affect horses and can even migrate through the blood vessels of the gut to the liver and skin, causing catastrophic damage.

Plants are also susceptible to helminth infection. Larvae can cause serious disruption to leaves and vascular system, compromising the plant's ability to photosynthesise and transport water and nutrients.

TABLE 10.4 Examples of helminth diseases in animals

HELMINTH	NAME OF DISEASE	FEATURES OF DISEASE
Enterobius vermicularis	Threadworm/pinworm (most common worm infection in Australia)	• Red and itchy anus • Irritability and behaviour changes • Lack of appetite • Restless sleep • Worms may be visible in faeces • Weight loss • Abdominal pain • Diarrhoea • Blood in faeces
Taenia solium (pigs) *Taenia saginata* (cattle)	Tapeworm	
Toxocara canis (dogs) *Toxascaris leonina* (cats) *Ascaris lumbricoides* (humans)	Roundworm	
Ancylostoma duodenale (humans) *Ancylostoma caninum* (dogs) *Unicnaria stenocephala* (dogs, cats, foxes)	Hookworm	

Parasitic arthropods

Arthropods are invertebrates that have an exoskeleton and a segmented body; they include insects and arachnids. Parasitic arthropods are ectoparasites, a leading cause of disease in humans, animals and plants. They may cause skin irritation, act as vectors for other pathogens and contribute to blood loss and concurrent infections. In some cases, ectoparasites have a devastating effect on organisms. Treatment generally involves chemicals.

Fleas

Fleas infest a variety of animals, including humans. They can transmit tapeworms in dogs (*Dipylidium caninum*) and cause allergic reactions to their saliva, which contains an antigenic protein. Flea allergy dermatitis is caused by hypersensitivity to fleas and is a common problem in dogs. It typically manifests as hair loss and skin redness along the top of the back, or 'hot spots', near the tail and other areas on the belly. Flea eggs inhabit the soil, cracks in floorboards and paving, and generally anywhere a household pet frequents. The eggs are sensitive to vibrations and carbon dioxide concentrations, and will often hatch in the thousands when a house's inhabitants return from holidays (Fig. 10.22).

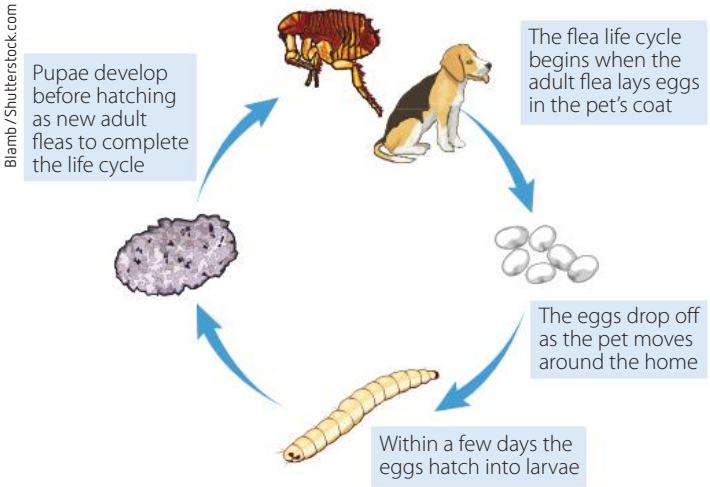

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FIGURE 10.22 The life cycle of the dog flea, *Ctenocephalides canis*

Ticks

Ticks are common ectoparasites of humans and animals. The paralysis tick *Ixodes holocyclus* (Fig. 10.23) is a problem in wet, densely vegetated areas, such as north-west Sydney. The tick injects a neurotoxin during feeding that causes progressive paralysis. Ticks can be extremely difficult to locate on a pet and their

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effects may last several weeks, even after they have been removed. Bandicoots and possums are natural hosts but are generally unaffected unless they carry large numbers of ticks. One of the first signs of tick paralysis in dogs is **dysphonia** (a strange bark) followed by **dysphagia** (difficulty swallowing), a staggering gait (**ataxia**) and potentially life-threatening paralysis.

FIGURE 10.23 The paralysis tick, *Ixodes holocyclus*

Lice

Lice may have biting or sucking mouthparts (Fig. 10.24a). They cause economic loss in the wool industry as itchy sheep scratch themselves on fence posts and damage their fleece. Sucking by lice can cause anaemia in infested animals. Head lice (*Pediculus humanus capitis*) are a common problem in schools and childcare centres due to close contact between children.

Mites

Mites are arachnids (Fig. 10.24b). They live mostly on animals but can also infest humans. Infestation with mites causes a condition known as **mange** (Fig. 10.3). Mites generally cause irritation, itchiness and redness of the skin. House dust mites feed on shed human skin flakes. They are responsible for many respiratory and skin problems in humans due to allergies.

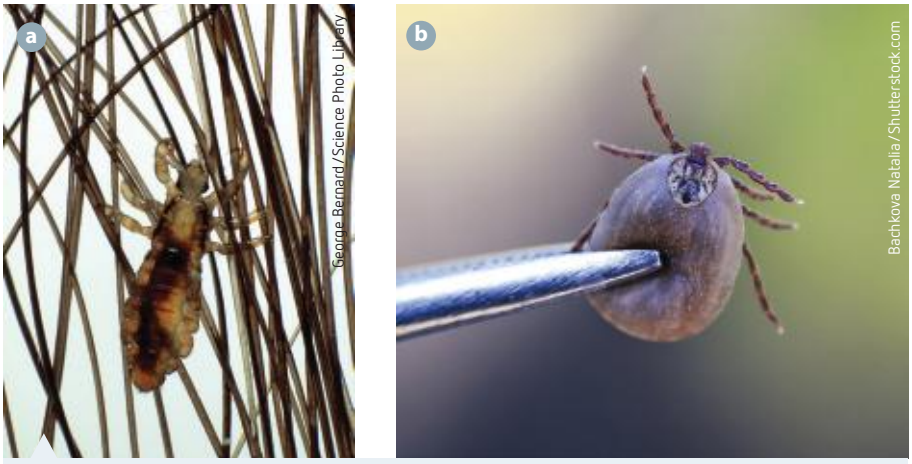

FIGURE 10.24 **a** Lice are insects; **b** mites are arachnids

TABLE 10.5 Examples of mites that infest animals and humans

MITE	DISEASE	FEATURES
Demodex canis (dogs)	Demodectic mange	• Overpopulation of the parasitic organism due to immune compromise of the host • Hair loss (alopecia) • Itching • Red skin • Crusting of skin
Sarcoptes scabei	Scabies	• Hair loss (alopecia) • Itching • Red skin • Crusting of skin
Ornithonyssus bursa	Bird mites	• Hair loss (alopecia) in humans • Itching • Red skin • Crusting of skin


Jon Ongkiebong/Alamy Stock Photo

FIGURE 10.25 Cattle bunch together when flies begin to bite them, in an attempt to avoid being bitten

Flies

The common housefly is found everywhere around the world. It is a non-biting fly but is capable of transmitting pathogens between hosts, including potentially dangerous gut bacteria such as *E. coli*. Some flies lay eggs in the damaged skin of their hosts. Fly strike in sheep is caused by the sheep blowfly (*Lucilia cuprina*) and is a devastating condition where the eggs hatch and the maggots burrow into the sheep's flesh and feed from it. Stable fly in horses (*Stomoxys calcitrans*) can lead to reduced weight gain in the horse because of its continual agitation caused by the flies biting. Large populations of flies around cattle yards and paddocks can cause severe stress to animals as they shake their heads and flap their ears in an attempt to rid themselves of the flies, even developing heat stroke in summer.

Mosquitoes

Worksheet
Diversity of pathogens

Mosquitoes can be considered biological parasites, because they benefit by having blood meals from their hosts. Mosquitoes also act as vectors, transferring pathogens from one human or animal to another. These pathogens include those that cause malaria, Ross River fever, yellow fever and Dengue fever. Zika virus, transmitted by mosquitoes, is an emerging concern around the world and is especially dangerous because of its effects on the unborn foetus. It was first reported outside Africa and Asia in 2007.

INVESTIGATION 10.1

A primary investigation of the comparative sizes of pathogenic cells

Pathogens vary widely in shape and size. With advances in technology, we are now able to see objects of astonishingly small sizes. The identification of pathogens is an important tool for a clinician who wishes to identify the causal agent of an infectious disease. One distinguishing feature that can be easily assessed under the light microscope is the comparative size of many cellular pathogens.

Refer to *Biology in Focus* Year 11, Chapter 2 to revise how to use a microscope.

AIM

To use a light microscope to examine and compare the sizes of a variety of cellular pathogens

MATERIALS

- light microscope
- prepared slides of protozoa, bacteria, moulds, yeasts
- oil immersion lens
- immersion oil

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RISK ASSESSMENT

Complete a risk assessment for your investigation.

WHAT IS A HAZARD?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?

METHOD

1. Observe prepared slides of yeasts, moulds and protozoa under low power and then high power.
2. Draw a diagram of each of type of cell. Label the cell wall, cell membrane, nucleus, cytoplasm and other structures you may see. Note the magnification.
3. Observe a prepared slide of bacterial cells.
4. Use an oil immersion objective lens to give a higher magnification (to observe the bacteria). Place a drop of immersion oil on the coverslip above the specimen and centre the oil immersion objective lens over the oil. Use the fine focus adjustment to obtain a clear image.
5. Draw a picture of one bacterial cell. Note the magnification.

RESULTS

Record your results in a table that includes:

- the cells recorded in order from smallest to largest
- a scientific diagram of each pathogen with a scale line to indicate size
- the magnification you used for each
- any similarities and differences noted between the different cellular pathogens.

DISCUSSION

1. Imagine that a doctor is in a remote village in a developing nation, investigating an outbreak of an infectious disease. Discuss the usefulness of the above method in this situation.
2. What kinds of information about the pathogen or the disease would the doctor *not* get from this type of investigation?
3. Evaluate the importance of keeping tissue samples free of contamination from the environment when preparing slides for this kind of investigation in the field.
4. What extra precautions would need to be taken by a person who is preparing their own slides of pathogens rather than using pre-prepared ones?

CONCLUSION

Write a summary statement on the usefulness of the light microscope in identifying pathogens by their comparative size.

KEY CONCEPTS

- Cellular pathogens include bacteria, fungi, protozoa and macro-organisms.
- Bacteria are prokaryotic, unicellular organisms that are mostly harmless in nature. Some are pathogenic to humans, animals and plants, and may directly destroy tissue or secrete toxins.
- Fungi may be unicellular or multicellular and are common causes of skin and lung diseases in humans and animals.
- Protozoa are unicellular eukaryotes that inhabit a wide range of ecosystems; some are pathogens.
- Macro-organisms such as worms (helminths) and arthropods (flies, mosquitoes, ticks, fleas, lice, mites) can be pathogens or can act as vectors and carry other pathogens.

Non-cellular pathogens


HECTOR RETAMAL / Getty Images

FIGURE 10.26 A rotavirus vaccine is available for infants.

In Sydney during the winter of 2017, an increased number of people presented to hospital emergency departments with severe vomiting and diarrhoea. Particularly hard hit were aged care facilities and childcare centres. The culprits were *Norovirus* and *Rotavirus*, which are spread by direct contact with an infected person. NSW Health advised people to wash their hands after visiting the toilet and before eating, to stop the spread of the virus. A rotavirus vaccine is available for children (Fig. 10.26).

Viruses

The word 'virus' is derived from a Latin word meaning 'slimy liquid or poison'. Viruses were first recognised as pathogens in the 1890s after the study of the tobacco mosaic virus. Viruses are non-cellular and have both living and non-living characteristics: they contain genetic material in the form of nucleic acids and are able to pass on hereditary information (a characteristic of living organisms), they are not composed of cells and they can be crystallised (a characteristic of non-living things). Viruses are not free living – they can only reproduce and metabolise in a host cell. A single viral particle is known as a virion.

Classification of viruses

Viruses are so small that they can only be viewed using an electron microscope. They consist of a protective protein coat (called a **capsid**) that encloses the genetic material, which may be DNA or RNA – this is the infectious part of the virus. Viruses that contain RNA are known as **retroviruses**. Some viruses also have a lipid membrane (envelope) that surrounds the capsid (Fig. 10.27).

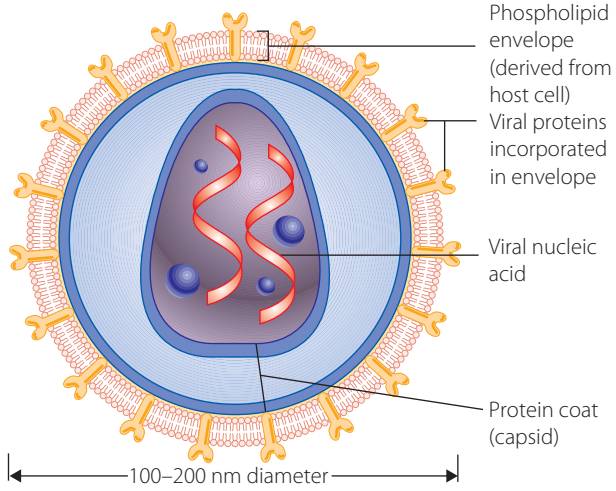

FIGURE 10.27 Generalised structure of a virus

Each virus has a particular type of structure. Some common structures are shown in Figure 10.28.

The viral protein coat contains chemicals that allow the virus to attach to the surface of the host cell. Once the virus has attached to a cell, it enters and takes over the cell's reproductive mechanisms, making many copies of itself. The host cell becomes so full of copies of the virus that it dies and bursts, releasing the new viruses, which repeat the replication process in other host cells (Fig. 10.29). Some viruses use the cell membrane of the host to form their own lipids and glycoproteins. **Bacteriophages** (viruses that invade bacterial cells) reproduce in the same way, but instead of entering the host cell they simply inject their genetic material into it.

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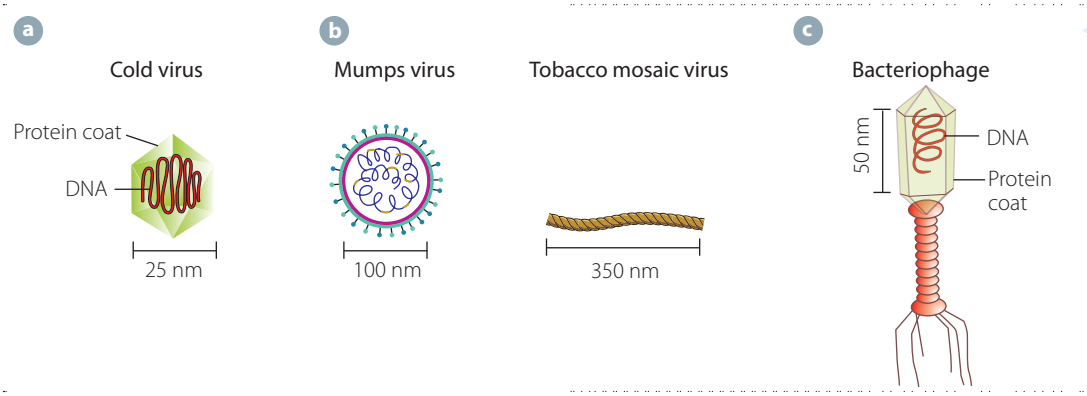

FIGURE 10.28 Structures of different viruses:
a icosahedral;
b spherical virion
c helical (filamentous);
d complex

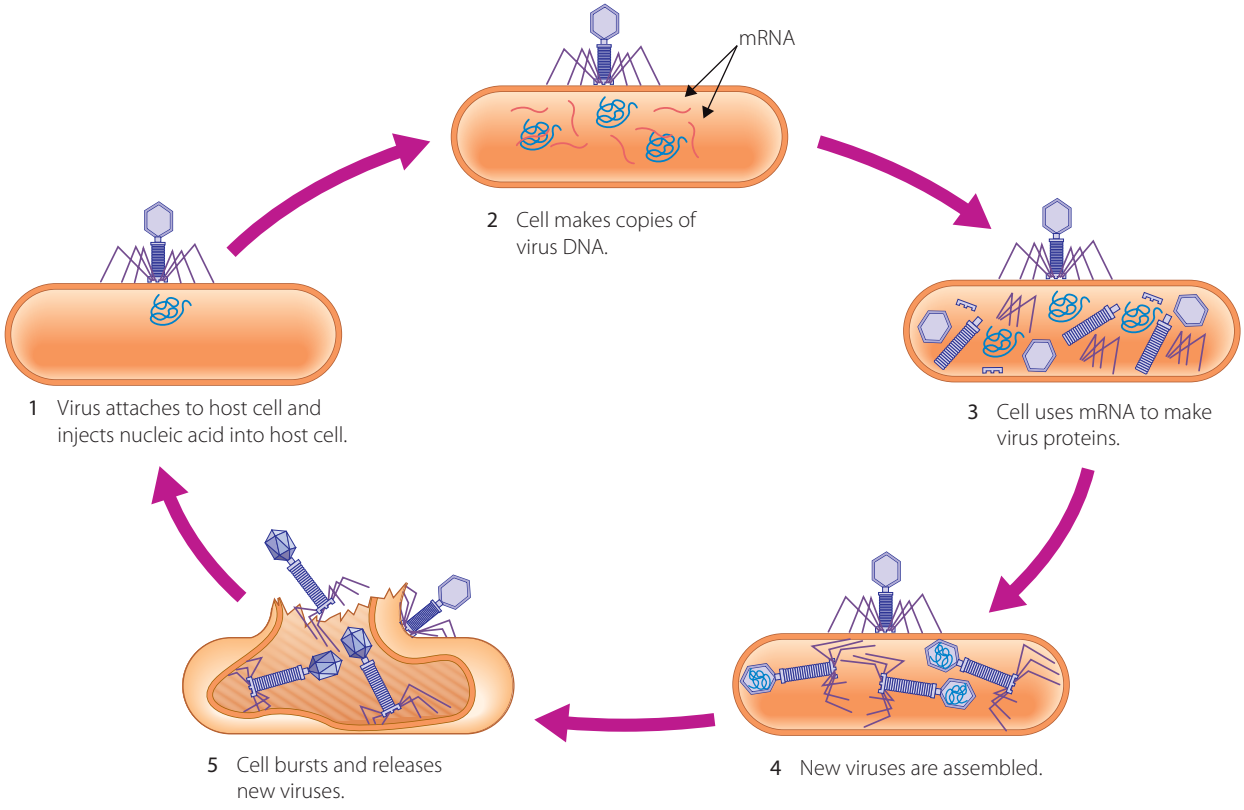

FIGURE 10.29 Replication of viruses

Many diseases are caused by viruses, including:

- influenza
- measles
- AIDS (caused by the human immunodeficiency virus, HIV)
- herpes
- glandular fever
- SARS (severe acute respiratory syndrome)
- tobacco mosaic virus disease in plants.

The treatment of viral diseases is difficult, as any attempt to kill the virus will also affect the host cells.

Prions: kuru

In the highlands of Papua New Guinea in the 1950s, in a tribe of people known as the Fore, researchers became aware of an unusual disease. It caused shivering and trembling, and often uncontrolled laughter, and rapidly progressed to death. At least 200 people a year had succumbed to this mysterious disease. Eventually the researchers linked this disease to the practice of eating people who had passed away. The brain was consumed in an attempt to spare the deceased the indignity of being eaten by worms. The pathogen was passed on in this way. The researchers won a Nobel Prize in Physiology or Medicine for identifying a completely new infectious agent – the prion. The disease was named kuru.

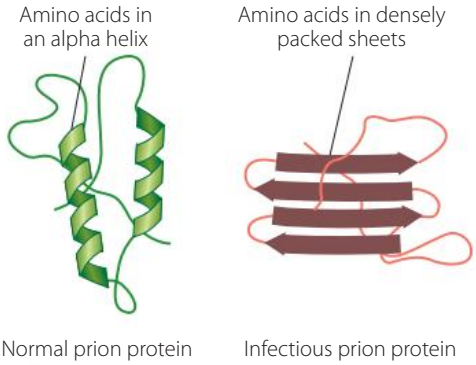

FIGURE 10.30 Normal prion proteins and pathogenic prion proteins have different shapes.

Classification of prions

A pathogenic prion is an abnormal protein that is capable of causing degenerative diseases of the nervous system. Unlike other types of pathogens, prions do not contain any genetic material (DNA or RNA). Prions are the smallest of all pathogens.

Disease-causing prion proteins have a different structure to normal prion proteins, which are found in the brain and spinal cord of humans and animals (Fig. 10.30). Pathogenic prions cause disease by inducing abnormal folding patterns in the normal proteins that they come in contact with (Fig. 10.31). The abnormal proteins are deposited within the central nervous system and other organs.

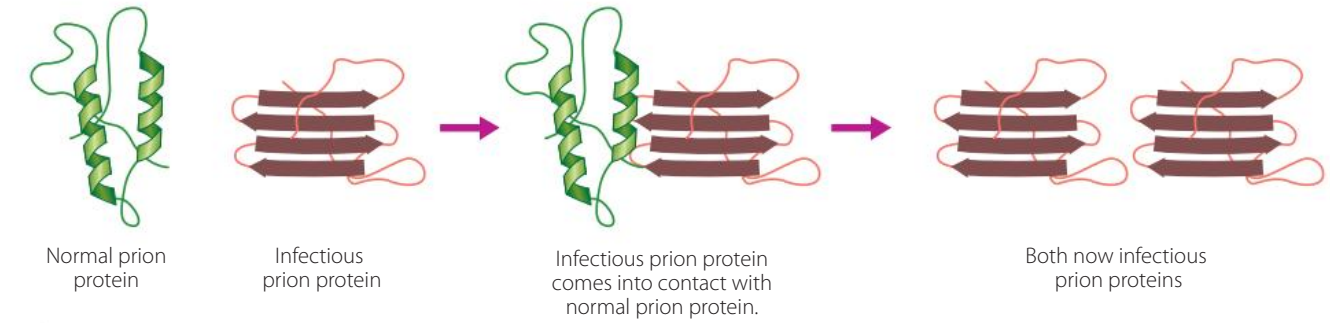

FIGURE 10.31 Conversion of normal prion proteins into infectious prion proteins

The diseases caused by pathogenic prions are known as transmissible spongiform encephalopathies (TSEs). They are called 'spongiform' diseases because the brain tissue of infected individuals is full of holes, resembling a sponge (Fig. 10.32). These diseases have a very long incubation period (5–20 years), but can progress rapidly once the first clinical signs appear. Other examples of TSEs are 'mad cow' disease or BSE (bovine spongiform encephalopathy) and CJD (Creutzfeldt-Jakob Disease).

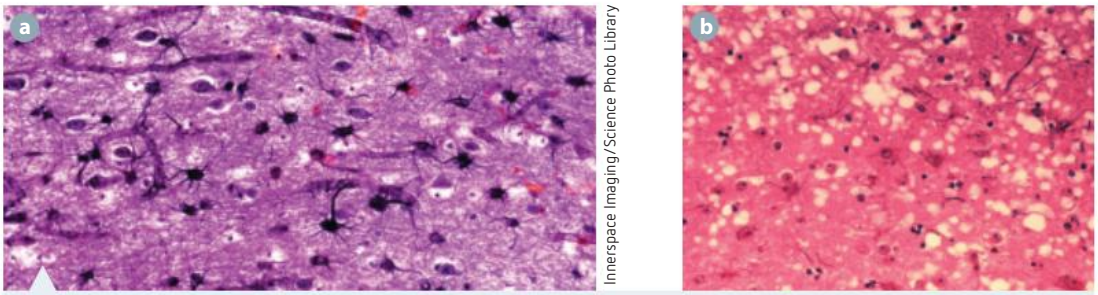

FIGURE 10.32 Changes in the brain caused by pathogenic prions: **a** normal brain tissue; **b** brain tissue affected by prion disease (the white spaces are holes)

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Transmission of prions and TSE diseases

Prions can be contracted in the following ways:

- ingesting tissue containing infectious prions, such as nervous and brain tissue
- as a result of surgery where contaminated equipment was used
- receiving contaminated growth hormone injections taken from infected animals
- receiving contaminated corneal transplants from previously infected organ donors
- inheriting the mutated gene that codes for the infectious prion
- spontaneous formation of infectious prions.

Diseases caused by prions

Diseases caused by prions include:

- Creutzfeldt-Jakob disease (CJD) in humans, a rapidly progressive and fatal brain disease
- variant CJD in humans (related to 'mad cow disease' or bovine spongiform encephalopathy, which occurs in cattle)
- chronic wasting disease of north American deer, elk and moose (Cervidae)
- scrapie in sheep and goats
- kuru in humans
- fatal familial insomnia – from the spontaneous transformation of protein in the brains of humans.

KEY CONCEPTS

- Non-cellular pathogens include viruses and prions.
- Viruses are obligate parasites.
- Viruses contain DNA or RNA in a protein coat.
- Viruses use the contents of the host cell to replicate their genetic material and create new virions.
- Prions consist of abnormally folded prion protein.
- Normal prion protein is found throughout the brain and spinal cord in humans and mammals.
- Disease-causing prions induce abnormal folding patterns when they come in contact with other prion proteins.
- Prion diseases have a long incubation period and are inevitably fatal once clinical signs occur.

CHECK YOUR UNDERSTANDING 10.1a

1. Are all bacteria pathogens? Justify your answer.
2. Anthrax spores can survive in dry conditions for decades. Find out what makes a spore so resistant to desiccation.
3. Describe the characteristics that could be used to distinguish between the following pathogens:
   - **a** bacteria and viruses
   - **b** fungi and protozoans
   - **c** prions and viruses
   - **d** bacteria and fungi
   - **e** protozoans and macroparasites.
4. When treating a patient for an infectious disease, a doctor would need to know whether the treatment was effective. How would a dead bacterium look different from a living bacterium under a light microscope? Would you need more information before you could tell whether a pathogen is dead, rather than just relying on its appearance? Explain your answer.
5. Identify the differences between the composition of the cell walls of bacteria, fungi and plants. Assess the usefulness of these differences when trying to identify the causal agent of an infectious disease.
6. Explain why coming into contact with a pathogen does not necessarily result in disease.
7. Define the following terms: virulence factors, incubation period.

8 Contrast the way in which a virus reproduces with the way prions propagate in infected brain tissue.

9 Choose two infectious diseases and briefly explain how each is commonly transmitted.

10 How would a microbe that is part of the normal microflora become a pathogen?

Modes of transmission of infectious disease

Transmission of infectious diseases involves the carrying or transfer of a pathogen from an infected host to a non-infected organism. The mode of transmission relates directly to the ability of the pathogen to survive outside a host cell. Many pathogens can persist outside the host in a state of dormancy. Pathogens may exist in **reservoirs**, either in the environment or in living hosts. For example, spore-forming bacteria such as anthrax can exist for years in a soil reservoir where the body fluids of an infected animal have seeped into the soil.

The human gut can act as a reservoir for pathogens. A human may be an active **carrier**, harbouring the disease in their own body, or a passive carrier, transmitting the pathogen from person to person on unwashed contaminated hands (in a hospital setting, for example). A famous historic example of an active carrier was Mary Mallon, also known as 'typhoid Mary', who was an asymptomatic carrier of typhoid fever (*Salmonella typhi*). Mary was a cook for many households in New York City between 1900 and 1915. During this time, she infected around 122 people, five of whom died.

In macroparasitic infections, there are often several hosts in the life cycle and therefore several different modes of transmission. The parasite must be transmitted between each host to reach sexual maturity. Each stage of the life cycle must be successfully completed and multiple transmissions occur. For example, the liver fluke (*Fasciola hepatica*) uses humans, sheep or cattle and snails as an intermediate host to complete its life cycle (Fig. 10.33).

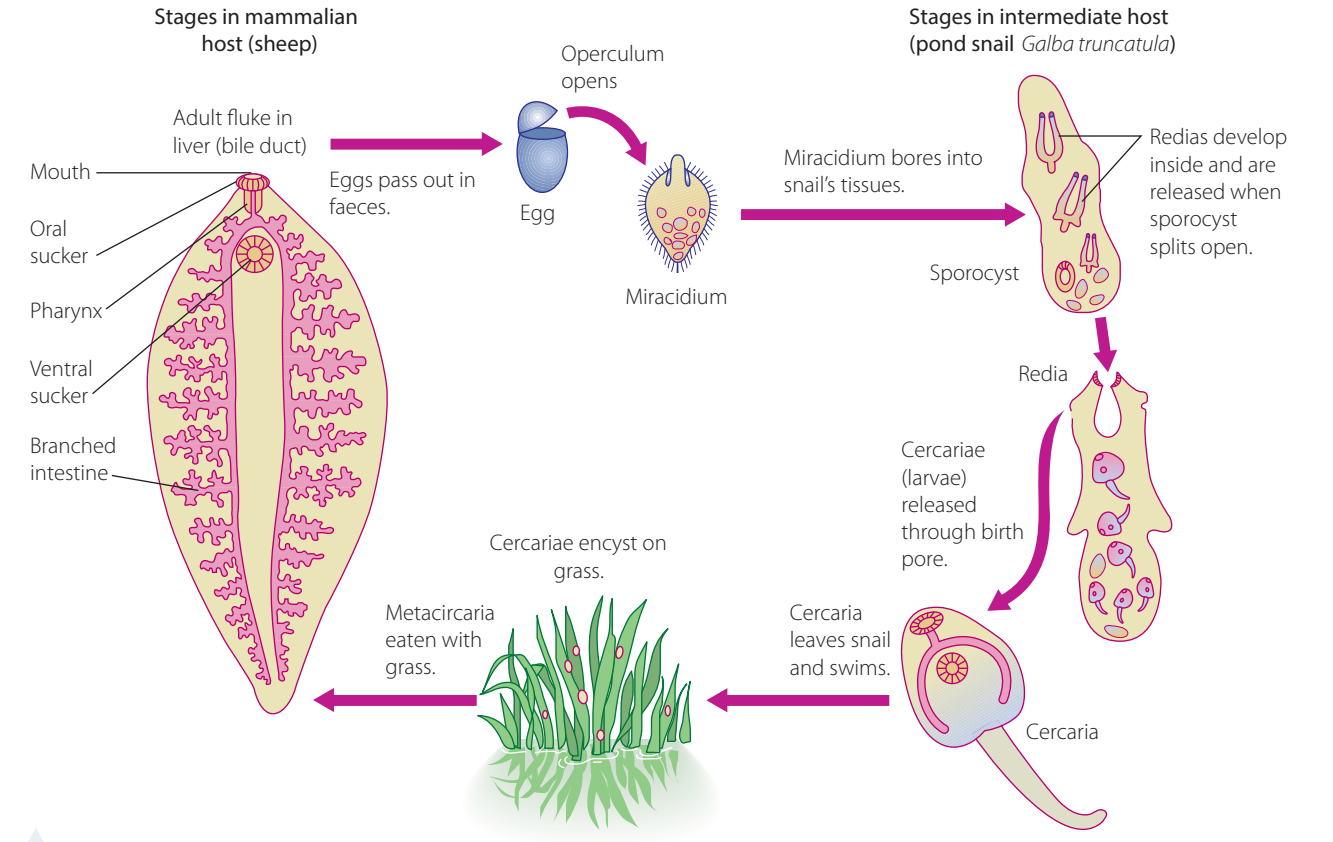

FIGURE 10.33 The complex, multistage life cycle of *Fasciola hepatica*, or liver fluke

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An understanding of transmission methods is critical to the control of disease outbreaks.

For a disease to spread between organisms, a 'chain of infection' must be present. This chain has three elements:

- a *host* that is susceptible to the disease
- a *pathogen* that is capable of causing the disease
- a *mode of transmission* – a way for the pathogen to get from host to host.

There are three modes of transmission, or ways that a pathogen can get from host to host:

- direct contact – transfer of the pathogen via exposure to infected skin or body secretions
- indirect contact – transfer of the pathogen to a new host via a non-living object
- vector transmission – transfer of the pathogen via another organism, such as an arthropod.

Direct contact

Transmission by direct contact occurs when there is physical contact between the host and a non-infected organism. Contact between organisms of the same generation, or between organisms that are not parent and child, is known as **horizontal transmission**. Contact between offspring and parent (for example, from mother to baby during childbirth) is known as **vertical transmission**. Physical contact includes:

- touching
- sexual contact
- kissing
- contact with nasal or oral secretions
- biting
- direct contact with any blood or other body fluids
- direct contact with wounds
- **prenatal** (before birth or during pregnancy) or **perinatal** (around the time of birth) transmission.

Diseases caused by direct contact include:

- skin infections, such as ringworm and impetigo
- cytomegalovirus (CMV) or glandular fever
- HIV/AIDS
- herpes simplex virus (HSV).

Indirect contact

Transmission by indirect contact occurs when the host and another organism have no direct contact with each other. Infection occurs from a reservoir created by the host outside itself, such as contaminated materials and surfaces or objects. A **fomite** is any object or substance that carries infection. Airborne diseases are of grave concern, as they are often the most difficult to control once an outbreak occurs. Infection may also occur via a vector.

Indirect means of transmission include:

- airborne transmission – coughing or sneezing (droplets can travel up to 8 metres through the air)
- touching an infected surface
- contaminated food or water
- infected surgical instruments that have not been **sterilised** correctly (surgical instruments are generally sterilised by exposure to saturated steam under pressure, in an **autoclave**)
- vectors such as mosquitoes, ticks and fleas. (See next section.)

Examples of diseases spread by indirect contact include:

- measles virus – from infected droplets (bodily fluids)
- gastroenteritis, caused by the bacterium *E. coli* – from contaminated food and water

- toxoplasmosis, caused by the protozoan *Toxoplasma gondii* – from infected cat droppings
- Legionnaires' disease, caused by over 50 species of *Legionella* bacterium – from contaminated water in water cooling towers
- influenza – exposure to droplets and other biological matter containing influenza virus particles.

Vector transmission

Vector transmission is a special case of indirect transmission of pathogens. It occurs through arthropods such as certain species of mosquitoes, sandflies, ticks, fleas and flies, or through infected aquatic snails. It usually involves a bite from an arthropod that is bloodsucking and transmits the pathogen during a meal, although in some cases, animals swallow the arthropod in the act of grooming themselves (for example, bot flies in horses, flea tapeworms in dogs). Sometimes infected plants and fungi are sources of pathogens and can act as vectors. Mammals such as fruit bats are vectors for the Hendra virus, and pigs may act as vectors for Menangle virus.

Every year, millions of people die from infection via a vector, and these diseases represent around 17% of all infectious diseases in humans. Transmission of these diseases is influenced by a complex association of environmental and social factors. For example, vector diseases are most common in warm, humid parts of the world where the conditions for insect survival and reproduction are favourable. Mosquitoes that transmit malaria (female *Anopheles*), for example, lay their eggs in water.

Diseases spread by vector transmission include:

- Chagas disease
- malaria
- dengue fever
- leishmaniasis (once considered **exotic**, now a reservoir in macropods)
- schistosomiasis
- onchocerchiasis
- canine and feline heartworm (*Dirofilaria immitis*)
- Hendra and Nipah viruses.

Case study: Equine influenza virus

Equine influenza (horse flu) is an exotic equine disease. If it became **enzootic** (endemic in an animal population), it would have a devastating effect on the Australian horse industry. In August 2007, an equine veterinarian reported several cases of sick horses in Centennial Park, Sydney. At the same time, breeding stallions from Japan at the Eastern Creek Quarantine Station were showing signs of equine influenza virus (EIV) infection after their importation. Laboratory tests confirmed an outbreak of EIV in both groups of horses. Certain protocols had already been set in place in anticipation of an outbreak, as per the Australian Veterinary Emergency Plan (AUSTVETPLAN).

Cause

Equine influenza virus is an *orthomyxovirus* that affects horses and donkeys. It does not infect humans. Equine influenza is caused by two main strains of EIV, known as equine-1 (H7N7) and equine-2 (H3N8). Symptoms include a fever (38.5°C or higher), watery nasal discharge, hacking cough, loss of appetite and muscle pain. Horses appear depressed and may have laboured breathing.

Transmission

Equine influenza is highly contagious. It is spread:

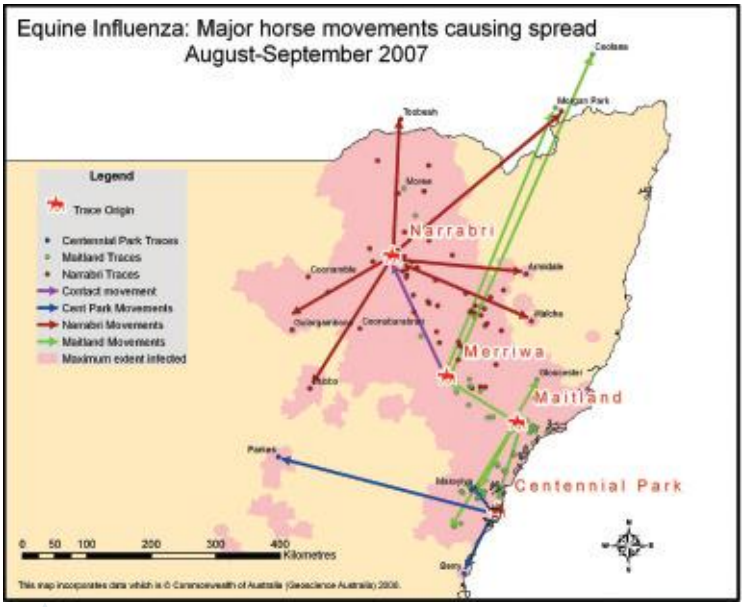
- directly between infected horses through nasal secretions and other body fluids
- indirectly through humans who carry the virus from an infected horse to other horses via contaminated shoes, clothing, grooming equipment, food and water buckets.

Management of the outbreak

Once a diagnosis of equine influenza was confirmed, the following measures were enacted.

- The NSW Chief Veterinary Officer imposed a state-wide lockdown on movement of all horses, which eventually became nation-wide, to prevent further spread of the virus.
- A management centre was set up to coordinate management at the disease control headquarters in Orange and Menangle, NSW.
- Horse properties were quarantined throughout NSW.
- The spread of the disease was mapped (Fig. 10.34), and by the end of August it was clear that the virus had spread to the NSW Central Coast and the Hunter Valley. Areas of high horse stocking density (such as Dubbo) experienced the fastest spread of the disease.

With the lockdown of horse movements and quarantine procedures, the outbreak was controlled by December 2007 (Fig. 10.35). Australia was declared free of the virus in early 2008.



Source: Moloney, Taragela and Buckley / NSW DPI Biosecurity

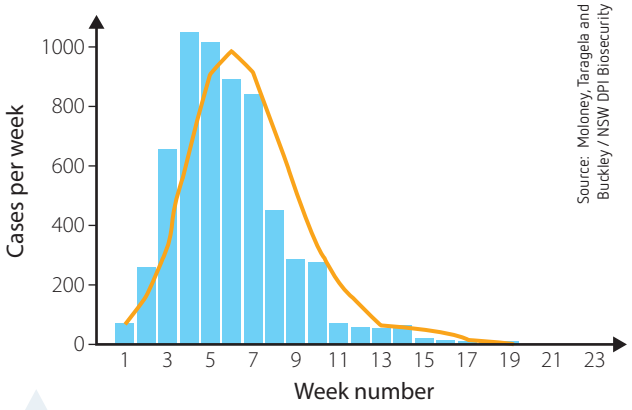
FIGURE 10.34 The movement of horses caused the equine influenza virus to spread.

Control of future outbreaks

The Australian Veterinary Association suggests that mass vaccination of Australian horses is not a justifiable option. The equine influenza virus mutates in the same way as human influenza viruses, and so vaccination would not confer immunity against new strains of the virus. Even vaccinated horses can still spread the virus indirectly. Vaccination may in fact delay detection of the disease.

There are a number of other ways in which future outbreaks of EIV can be controlled.

- Restrict the importation of live horses to those from approved countries.
- Subject imported horses to strict biosecurity measures, including: quarantining the horses in their own country for 14 days prior to export; and quarantining them upon arrival, in a post-entry quarantine facility in Victoria for a minimum of 14 days.
- Public education, particularly for those working in the horse industry, is vital for early detection.
- Provide biosecurity training for all involved in the importation of horses into Australia, including grooms, truck drivers, cleaners and airline staff.



Source: Moloney, Taragela and Buckley / NSW DPI Biosecurity

FIGURE 10.35 The weekly number of new cases of EIV peaked and then declined steadily in 2017.

INVESTIGATION 10.2

Secondary-source investigation of transmission of a disease during an epidemic

Use the equine influenza case study described in this section to guide your own research on an infectious disease outbreak of your choice.

1 In writing up your investigation, include the following parts:

- **a** **Title** for your investigation
- **b** **Introduction** – the background to the outbreak. What were the events that led to it? What are some features of the pathogen and the hosts? Give some context to the outbreak by describing where it began, how it was first noticed, and so on.
- **c** **Aim** – write an aim for your investigation.
- **d** **Hypothesis** – this is an opportunity to dig a little deeper into an area of interest by formulating a hypothesis about a particular disease outbreak.
- **e** **Method** – choose an infectious disease that you wish to study. It may be a disease of humans, animals or plants. When considering which disease to study, you should take into account your interests and whether the disease can easily be studied according to the parameters given. Suggestions include (but are not limited to): measles, whooping cough, swine or avian influenza, Panama disease of bananas, hepatitis B, HIV/AIDS, Ebola, Hantavirus, SARS (severe acute respiratory syndrome), Hendra virus and variant Creutzfeldt-Jakob disease. From the secondary sources that you consult, identify the relevant material you need to describe that relates to the aspects of your chosen infectious disease.
- **f** **Resources** – a complete reference list should be provided in the style recommended by your teacher. Remember to use a number of different sources, including the Internet, books (such as medical reference material) and journals, and always assess the accuracy and reliability of these sources using the CRAAP method.

Refer to page 10 to review the CRAAP method.

- **g** **Communication** – present your information in a way that is considered suitable by your teacher. For example, you may like to design a fact sheet that describes information about your chosen infectious disease according to the areas listed above. Other possibilities include a multimedia presentation, video presentation, dramatic piece, detailed poster, or any other creative means. Brainstorm this with your class. You may like to work individually or as a group, in which case you need to organise the group members and assign roles. Make sure you use headings for each of the areas and include pictures/illustrations. Refer to the case study on equine influenza as a guide to the type of information that is required. You must include in your presentation:
  - **background** to the outbreak
  - **cause** – identify the specific pathogen and clinical signs of the disease
  - factors affecting **transmission** of the pathogen
  - **prevention and control** of the spread of the pathogen. Control refers to the strategies employed to keep the incidence of the disease to a minimum in the population. Control measures are aimed at eliminating the source of the infection, preventing the transmission of infection and protecting susceptible people. Prevention refers to strategies that can be used to stop the occurrence of the disease in individuals
  - **management/prevention** of future outbreaks of this pathogen. Examples are: isolation of the infected individual, interruption of the life cycle of the pathogen, immunisation programs, prevention of food and water contamination, and education programs.
- **h** **Discussion** – a general discussion of your findings that identifies major trends in your research. Also include some of the difficulties you faced in finding reliable data and some of the ways you overcame this.
- **i** **Conclusion** – refer to your aim (and hypothesis).

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Identifying microbes in food or water

Microbes are all around us – they are on and in our bodies, in the air, on the surfaces around us, in water and in food. Not all microbes cause disease; in fact, some have little or no effect on us, and others are even beneficial. Sometimes food and water can become contaminated by pathogens. These commonly cause symptoms such as vomiting and diarrhoea but occasionally can lead to more severe consequences, such as kidney failure and even death. Detection of microbes in food and water is not possible with the naked eye. However, many microbes, such as fungi and bacteria, cluster together when given suitable conditions for reproduction and growth, including moisture, nutrients and warmth. The clusters are known as colonies, and can be seen without the use of a microscope (Fig. 10.36).

All these conditions can be provided with the use of nutrient agar plates and an incubator set at approximately 30°C. Agar is a jelly-like substance derived from seaweed. It is dissolved in water, and nutrients suitable for microbial growth are added before it sets. Different pathogens require different nutrient agars. For example, streptococci bacteria often require a nutrient medium enriched with sheep blood, to grow well.

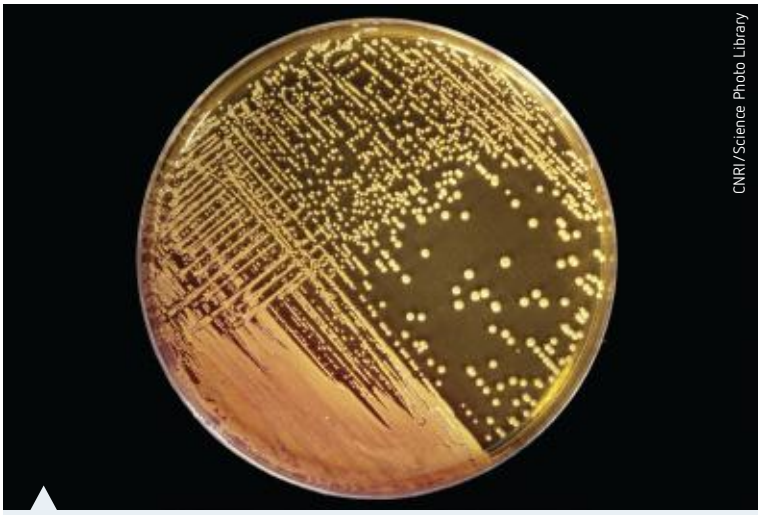

FIGURE 10.36 Bacterial colonies growing on a nutrient agar plate

The features of a colony can be used to help identify the specific pathogen. Bacterial colonies tend to be smooth, glossy and coloured, whereas fungal colonies are furry and large. Bacterial colonies can be identified according to their:

- colour
- margin
- form (basic shape)
- elevation (shape of the cross-section)
- surface features (smooth, dull, wrinkled) (Fig. 10.37).

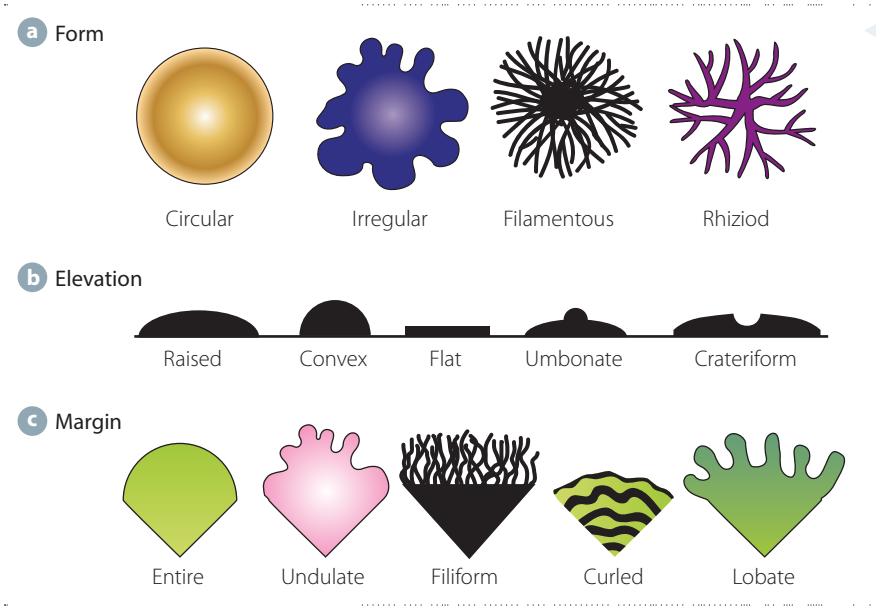

FIGURE 10.37 Different types of bacterial colonies can be identified, based on their **a** form, **b** elevation and **c** margin.

INVESTIGATION 10.3

Primary investigation into the microbial testing of food and water samples

Information and communication technology capability

Sterile technique is extremely important during this investigation. Read the worksheet, *The importance of sterile technique*, thoroughly before you start this investigation.

Worksheet
The importance of sterile technique

AIM

To design and conduct a practical investigation relating to the microbial testing of water or food samples

HYPOTHESIS

Your hypothesis should be in the form of a general statement. It should include both dependent and independent variables.

MATERIALS

Provide a numbered list of all equipment you use in this experiment. This includes all the personal protective equipment (PPE) you use, such as gloves, lab coats and safety goggles.

Weblink
Risk Assess

RISK ASSESSMENT

You must carry out a risk assessment that shows the hazard, what could happen and what procedures are in place to minimise the hazard. Use of an online risk assessment website is highly recommended. (See the weblink.)

RISK ASSESSMENT

WHAT IS THE HAZARD?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?

- Once the Petri dish has been sealed, it must not be opened again.
- All equipment that is used should be autoclaved. Reusable equipment can then be washed and Petri dishes disposed of in the correct manner.
- Always wash your hands thoroughly before leaving the laboratory.

METHOD

When planning the method, you must ensure that your investigation is a 'fair' test. You should consider the following:

- A *control* should be used to ensure the validity of the experiment. A control is designed to show what would happen without the presence of the variable being tested. The experiment should also be designed to test what it sets out to test.
- Identify the *independent variable* – the variable that is being tested in the experiment and the one that you change.
- Also identify the *dependent variable* – the one that you measure/observe and record in your results.
- All other variables in your experiment must be controlled.
- Carry out a risk assessment that shows the hazard, what could happen and what procedures are in place to minimise the hazard.
- Decide whether you are going to study food or water, and the number and sources of test samples. Try to use a variety of sources. If you are going to use food in this investigation, mash each sample up in a test tube with 2 mL of distilled water. Include instructions on how to inoculate your agar plates with the food or water samples.

The syllabus states that you are to identify the microbes present in *food* or *water* – it is not sufficient that the agar plates are merely exposed to air in different environments.

RESULTS

Draw up an appropriate table in which to record your results. This may include labelled photographs of the agar plates before and after inoculation with the test substance.

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DISCUSSION

In your discussion, answer the following questions.

1. Justify and evaluate the use of a control in this experiment.
2. In this experiment, identify the:
   - **a** independent variable
   - **b** dependent variable
   - **c** controlled variables.
3. Describe some of the procedures used to prevent the contamination of samples. How could these procedures be improved?
4. Evaluate your risk assessment. Was it adequate preparation for the investigation?
5. Compare the number and types of microbial colonies present in each sample.
6. Identify which sample was the most contaminated and suggest a reason for this.

CONCLUSION

This should answer the question asked in the aim. State the relationship between the two variables you tested.

EXTENSION

Applications of microbiological techniques in the food industry.

Use online resources to research the following:

- **a** What results should you expect from your investigation?
- **b** Compare the expected results with your actual results, and explain any anomalies in terms of your technique, equipment or experimental method.
- **c** How are these tests applied in the food industry? Find out about their application in the testing of foods for consumption by humans. How could these tests be applied to ensuring safe drinking water for urban and rural populations?

KEY CONCEPTS

For the transmission of an infectious disease to occur, there must be a:

- host that is susceptible to the disease.
- pathogen present that is capable of causing the disease
- way for the pathogen to get from host to host (mode of transmission).

There are three ways that a pathogen gets from host to host:

- direct contact – transfer of the pathogen via exposure to infected skin or body secretions
- indirect contact – transfer of the pathogen to a new host via a non-living object
- vector transmission – transfer of the pathogen via another organism such as an arthropod.

CHECK YOUR UNDERSTANDING 10.1b

1. How can a bacterial pathogen be transmitted from a carrier to a host?
2. Can *any* mosquito transmit malaria between humans?
3. Contrast active and passive carriers of diseases.
4. Use an example to show that transmission of a pathogen to the final host may involve several stages.
5. What three elements of the 'chain of infection' are necessary for disease transmission to take place?
6. How can hydatid tapeworms be transmitted from animals to humans?
7. Briefly outline, using examples, two ways in which pathogens may be transferred:
   - **a** directly
   - **b** indirectly
   - **c** by means of a vector.

8 Outline the importance of conducting a risk assessment when working with biological material.

9 Justify the technique of holding the lid of an agar plate at an angle over the plate when inoculating with a potential pathogen.

10 Identify some of the biosecurity controls in place for importing a live animal.

10.2 Robert Koch and Louis Pasteur

In the second half of the 19th century, there was a revolution in microbiology, primarily due to the research of Louis Pasteur and Robert Koch. Working separately, Pasteur and Koch were able to make an invaluable contribution to our understanding of infectious disease. Although they each used aspects of the other's work in their own research, the degree of collaboration and communication was minimal, as they did not get along with each other.

Historical understanding of the causes of disease

Prior to the work of Koch, Pasteur and others, the cause of disease and decay was explained by the theory of spontaneous generation. This involved the idea that life, such as maggots present in rotting flesh, arose spontaneously from non-living things. Early scientists Redi and Spallanzani performed experiments that showed that some form of living matter had to be present before other living things could appear. Their work was not widely accepted, however, and the theory of spontaneous generation remained the most widely believed explanation for disease and decay. Subsequently, Pasteur was able to disprove this theory and establish the 'germ theory of disease'. This theory states that germs (microbes) cause disease and that all micro-organisms come from pre-existing micro-organisms.

Koch's postulates

Robert Koch (1843–1910) (Fig. 10.38) was born in Germany and obtained his medical degree from Göttingen University. Koch made many contributions to the field of microbiology and the understanding of infectious disease. He was an expert on bacteriological techniques, and many of his techniques are still used today.

Koch developed the agar plate technique for growing micro-organisms, and used it to culture the isolated anthrax bacillus.

He also carried out an extensive study of the anthrax bacillus. He examined the blood of sheep that had died from anthrax and identified rod-shaped bacteria, which he isolated and grew in cultures. These cultured bacteria were then injected into healthy sheep, which subsequently developed anthrax. He repeatedly showed that the anthrax spores he had obtained from the pure cultures he had grown could cause the disease in other animals and kill them. These experiments added further weight to the germ theory of disease, as they showed that a micro-organism grown outside the body caused a disease.

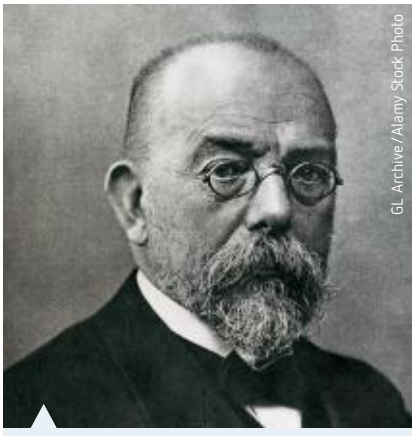
GL Archive / Alamy Stock Photo

FIGURE 10.38 Robert Koch

From this research, Koch determined that each disease is caused by a specific micro-organism. The principles he used to identify the specific micro-organism that was responsible for a disease came to be known as Koch's postulates (Fig. 10.39) and are still in use today to identify the specific micro-organism that causes a particular disease.

One of Koch's subsequent major breakthroughs was the discovery of the bacterium responsible for tuberculosis, *Mycobacterium tuberculosis*. He was also responsible for identifying the bacterium responsible for causing cholera. He travelled extensively in the latter part of his career, to study diseases such as bubonic plague and African sleeping sickness.

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The criteria that must be met to determine whether a particular micro-organism is responsible for causing a disease are known as **Koch's postulates** (Fig. 10.39) and are given below.

1. The same micro-organism must be present in every diseased host.
2. The micro-organism must be isolated and cultured in the laboratory and accurately described and recorded.
3. When a sample of the pure culture is inoculated into a healthy host, this host must develop the same symptoms as the original host.
4. The micro-organism must be able to be isolated from the second host and cultured and identified as the same as the original species.

Worksheet
Koch's postulates

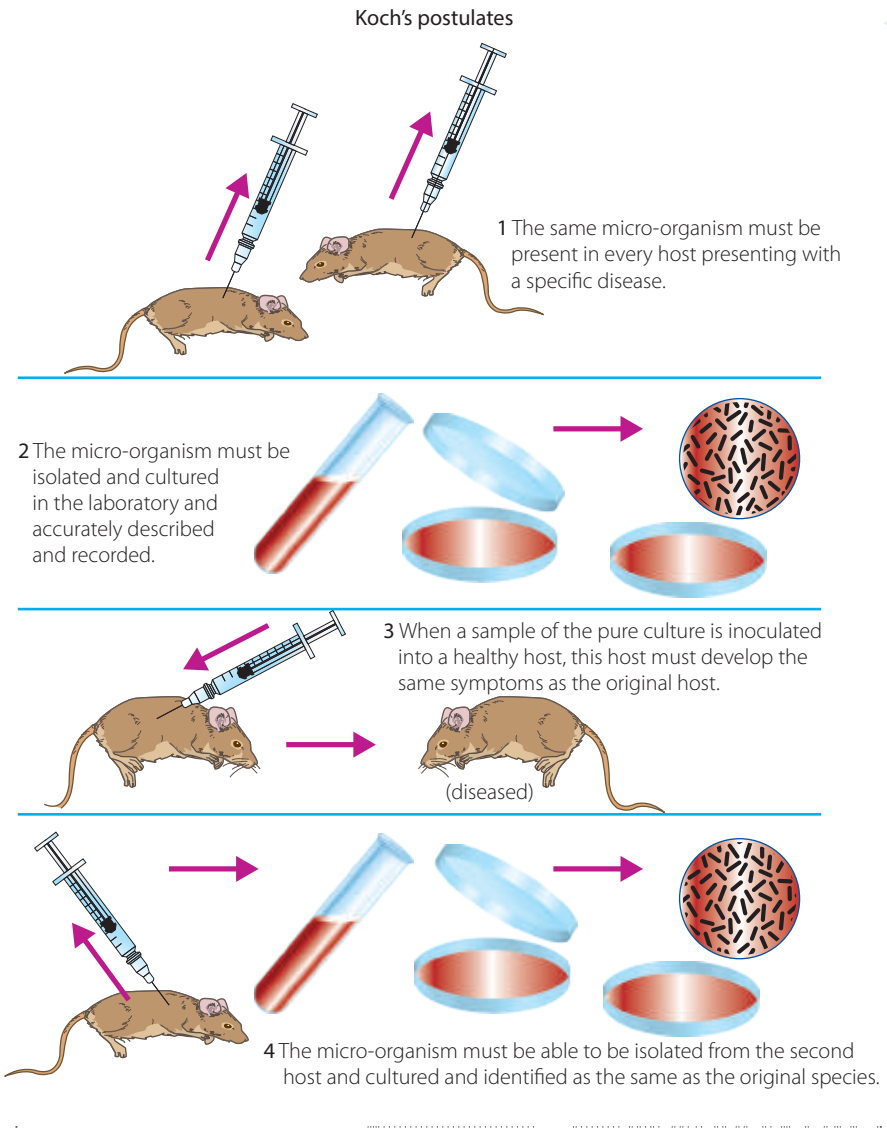

FIGURE 10.39 Koch's postulates

Pasteur's experiments on microbial contamination

Louis Pasteur (1822–1895) (Fig. 10.40) was born in France. Although not his original area of study, he was credited with creating the science of microbiology and made invaluable contributions to the understanding of infectious disease. At the request of an industrialist called Bigo, Pasteur studied the fermentation of beet juice and found that the process was due to the presence of living organisms – microbes called yeasts.


Science History Images/Alamy Stock Photo

FIGURE 10.40 Louis Pasteur

Pasteur also found that other micro-organisms were responsible for the souring of the alcohol produced. He was instrumental in finding that micro-organisms (bacteria) were the cause of wine, beer and vinegar spoilage. He also discovered that the solution to the wine and vinegar problem was to heat these solutions long enough to kill the contaminating bacteria that were present after fermentation. This was the beginning of the widely used process of **pasteurisation**, which is still used today to ensure that products such as milk are free of disease-causing micro-organisms and are suitable to drink. This process was initially used in milk production to destroy the tuberculosis bacterium.

Pasteur also found that the rotting of food was due to the activity of living organisms. After further investigation, he joined the debate over spontaneous generation, refuting this theory and proposing the germ theory of disease. He carried out his famous 'swan-necked flask' experiments to gain evidence to support his theory.

These experiments involved using flasks that had long drawn-out necks (like those of swans) that were not sealed (Fig. 10.41). Meat broth was boiled in the flasks and, as they cooled, the air was drawn in from outside. Any micro-organisms present in the air did not reach the broth, as they were trapped in the narrow neck and the curve of the glass. No bacterial or fungal growth was observed in these flasks. Bacterial growth occurred if the curve of the flask was broken off and the contents of the flask exposed to the air. Furthermore, tipping a flask to allow the solution in it to reach the curve where the micro-organisms were trapped resulted in bacterial growth occurring. This added further evidence to discredit the theory of spontaneous generation. It supported the hypothesis that the organisms that contaminated the broth and caused it to decay must be carried in the air and not be spontaneously generated.


Science & Society Picture Library/Getty Images

FIGURE 10.41 Pasteur's swan-necked flask

Pasteur's flasks are on display at the Pasteur Institute in Paris and, after more than 150 years, the broth in the swan-necked flasks is still free of bacterial growth. This classic experiment carried out by Pasteur demonstrates how theories in science can be disproved.

Pasteur also uncovered the relationship between micro-organisms and disease. As specific bacteria became associated with specific diseases, the spontaneous generation theory became less widely supported and the germ theory of disease grew more widely accepted. Pasteur also discovered the cause of silkworm disease and devised a test that allowed the selection of healthy eggs, saving the silkworm industry from potential disaster.

The work of Pasteur in the field of fermentation is also dealt with in Chapter 8.

Pasteur investigated the cause of anthrax and, after input from the experiments of Koch, determined that animals were contracting the disease even though they had had no known contact with animals suffering from the disease. This was due to spores from the carcasses of animals that had died from the disease. These carcasses had been buried in fields that were being grazed by healthy animals.

In studying fowl cholera, Pasteur also developed a way to attenuate, or weaken, bacteria so that when they are introduced into a host they can cause the body to be ready to recognise the real infection. He produced a vaccine that prevented chickens from developing chicken cholera, and then extended this work and developed a vaccine for anthrax. His detractors were not convinced of the effectiveness of the vaccine and he was challenged to carry out a public field test. This trial was successful, as all animals that were given the vaccine before being exposed to anthrax survived, and the animals that were not given the vaccine died. The vaccine was found to be effective and its use was widely adopted. Pasteur went on to develop vaccines for a number of other diseases, such as rabies, where he used the vaccine on humans for the first time.

Pasteur had established the principle of immunity and provided an effective way to prevent infectious disease.

Worksheet
Louis Pasteur and the germ theory of disease

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INVESTIGATION 10.4

Primary investigation to model Pasteur's swan-necked flask experiment

You are going to plan and set up an investigation to *model* Pasteur's famous experiment. When using a model it is important to identify what the different parts of your model represent in relation to the real thing.

You will be using beef stock cubes to make a broth similar to the broth Pasteur used. Conical flasks will replace Pasteur's balloon flasks, and glass tubing bent into an S-shape will be used to replace the swan-necked flask.

AIM

Write an aim for your investigation.

HYPOTHESIS

Write a suitable hypothesis for your investigation.

MATERIALS

Construct a materials list for your investigation. Make sure you identify the quantities of each material that you need.

RISK ASSESSMENT

Complete a risk assessment for your investigation.

WHAT IS THE HAZARD?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?

METHOD

Write a method to test your hypothesis.

RESULTS

Record your observations in a carefully planned table.

DISCUSSION

Discuss your results, paying particular attention to:

- your observations of each flask
- how well you ensured the accuracy, reliability and validity of your experimental design and how you could improve your experiment if applicable
- how this model is similar to and different from the actual experiments performed by Pasteur.

CONCLUSION

Summarise your findings about the conditions needed for transmission of pathogens.

KEY CONCEPTS

- Robert Koch and Louis Pasteur increased our understanding of the nature of infectious disease.
- Koch developed the agar plate technique for culturing microbes.
- Koch's work on anthrax, cholera, bubonic plague and tuberculosis contributed to management strategies for these diseases.
- Koch developed *postulates* to guide scientists in determining the causal pathogen for a disease.
- Koch showed that *specific* infectious diseases are caused by *specific* pathogens.
- Pasteur is credited with creating the science of microbiology, through rigorous experimentation.
- Pasteur identified microbes as the agents responsible for spoilage during the production of wine, beer and vinegar, leading to the development of pasteurisation.
- Pasteur's germ theory of disease was supported by his swan-necked flask experiment.
- Pasteur's work contributed to the development of vaccines for diseases such as fowl cholera, based on the principle of immunity.

CHECK YOUR UNDERSTANDING 10.2

1. In what ways were the causes of disease identified by Pasteur and Koch?
2. How did these scientists contribute to our understanding of the transmission of disease?
3. State the germ theory of disease. What previous theory does this attempt to disprove?
4. Briefly describe the contributions of Louis Pasteur to our understanding of infectious disease.
5. What are Koch's postulates? How did these contribute to our understanding of infectious disease?
6. Analyse Pasteur's swan-necked flask experiment as an example of the scientific method.
7. Justify, using a named experiment, Pasteur's claim that introduction of an attenuated pathogen can offer protection against subsequent infection.
8. How are Koch's postulates applied to emerging disease outbreaks today?
9. How would contamination of glassware have affect the outcome of the swan-necked flask experiment?
10. Were Koch's experiments on sheep ethical by today's standards?

10.3 Causes and effects of disease in agricultural production

Agriculture is a form of **primary industry** that involves the cultivation of crops and pastures and the rearing of animals to provide meat, milk, fibres and other products for humans. It was once said that Australia 'rode on the sheep's back' due to the enormous economic success of our wool industry.

The importance of agriculture in Australia

Because Australia is in the unique position of being isolated from the rest of the world, Australian agriculture is relatively free of many of the infectious diseases that affect animals and plants in other countries. This makes our agricultural products highly sought after around the world. Stringent biosecurity measures are in place to reduce the likelihood of disease transmission from pathogens. The introduction of new plant diseases could potentially devastate the horticultural industry, as well as forestry and agriculture.

Australia's main agricultural export products are listed in Table 10.6.

TABLE 10.6 Australia's top 10 agricultural exports (by value) in 2015

MAJOR AGRICULTURAL EXPORT PRODUCTS	VALUE (A$ m)	SHARE OF TOTAL (%)
Beef	9269	19.9
Wheat	5812	12.5
Meat (excluding beef)	3738	8.0
Wool and other animal hair	2911	6.2
Alcoholic beverages	2387	5.1
Vegetables	1931	4.1
Live animals (excluding seafood)	1896	4.0
Fruit and nuts	1805	3.9
Sugars, molasses and honey	1783	3.8
Barley	1740	3.7
Total of all exports	*44657*	

Source: Department of Foreign Affairs and Trade CC BY 3.0 AU license (https://creativecommons.org/licenses/by/3.0/au/)

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Infectious diseases in Australian agriculture

Two types of plant and animal diseases are of concern in agriculture in Australia:

- *endemic diseases* (diseases consistently present within a country or region) such as bovine Johne's disease in cattle, sheep and goats, anthrax in sheep and cattle, and footrot in sheep
- *exotic* (introduced) *diseases* such as foot and mouth disease, avian influenza (H5N1), bovine tuberculosis, equine influenza, and virulent Newcastle disease in domestic poultry and wild birds.

A complex interplay of three factors may contribute to the development of infectious disease in organisms of agricultural importance (Fig. 10.42):

- host factors – susceptibility to disease, access to pathogen, concurrent disease or poor nutrition leading to weakened immune response, drought and heatwave stress on the host
- pathogen factors – the pathogen's availability, its ability to transfer between hosts, as well as virulence factors including adhesion and invasion of host tissues, and successful establishment inside host tissues
- environmental factors – overcrowding and lack of hygiene leading to a build-up of wastes, which provide a suitable environment for pathogen reservoirs; a favourable environment within the host for pathogens to establish and cause disease.

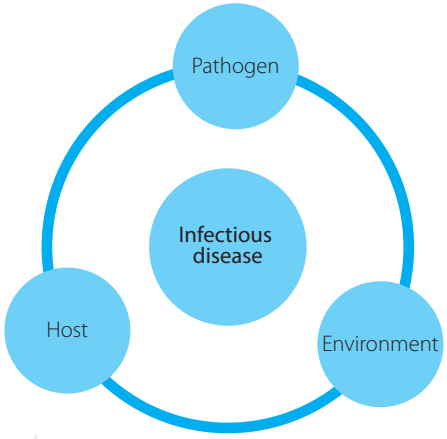

FIGURE 10.42 Factors affecting the incidence of infectious disease in agriculture

Case study: Footrot in sheep

Footrot is an infectious disease of the hooves of sheep, goats and cattle, caused by the pathogenic bacterium *Dichelobacter nodosus*. It causes painful abscesses between the toes (Fig. 10.43), lameness and weight loss, as grazing is affected.

FIGURE 10.43 Typical appearance of a sheep's foot infected with footrot

An outbreak of footrot on a farm depends on a number of factors being present.

- Pathogen factors – *Dichelobacter nodosus* must be present for footrot to occur.
- Environmental factors – the bacterium will only survive in soil outside the host for a maximum of 4 days. Pastures that are long, dense and wet aid in pathogen survival and transfer. Temperature is important: warm weather favours growth of the bacteria.
- Host factors – dry feet with intact tissues are not infected. Reservoirs of bacteria may form in individual animals' feet for years. Some dermatitis between the toes must already be present for bacteria to invade and establish an infection. Overgrown hooves provide a suitable environment for the bacteria.

Worksheet
The risk of infectious disease in agriculture

Factors contributing to the risk of infectious disease

Some of the factors that contribute to the increased risk of infectious disease in Australian agricultural production are outlined below.

- Increased mobility of human populations
  - Travellers, imported livestock and plants can carry infectious disease into Australia.
  - Both cellular and non-cellular pathogens can form a reservoir in food, soil and seeds on shoes, and in infected animals and animal products that are moved from one area to another by humans.
- Rise of intensive and industrial-type agriculture
  - The increase in the world's population has seen a change in the style of livestock production from extensive pastoral systems to intensive **feedlots**. Feedlots carry a higher risk of disease outbreak due to the higher **stocking densities** of animals. The closer animals are housed together, the more rapidly a pathogen can spread from animal to animal or plant to plant.
- Changing patterns of land use
  - **Deforestation** and irrigation practices may change the distribution of insects.
  - Loss of habitat can bring bats into closer proximity to human and horse populations (Hendra and Nipah viruses).
- Climate change
  - Distribution and abundance of insect vectors may change.
  - Changes in rainfall patterns may favour the formation of reservoirs of pathogens in soil, plants and insects.
  - Changes in ecosystems can change availability of nutrients to plants and animals and reduce immune responses to pathogens.
- Antimicrobial resistance
  - Antimicrobials are used to treat infections in livestock (e.g. mastitis in dairy cattle, which is an infection of the udder with environmental and faecal bacteria). The **'off-label' use** (use in a way that has not been officially approved) or overuse of antibiotics on farms hastens the development of antimicrobial resistance due to rapid natural selection of resistant bacteria. Antibiotics are also sprayed in orchards to treat plant infections. Antibiotic-resistant bacteria may be transferred to humans through direct contact with animals, consumption of their meat or transfer of genes between animal bacteria and human pathogens. If this happens, common bacterial infections may no longer be able to be treated with antibiotics.
  - Antimicrobials are used whenever animals are housed in high-density situations. The close proximity of animals facilitates easier transfer of pathogens during a disease outbreak. In this situation, animals are more exposed to their own waste products, which harbour bacteria. As well as being used to prevent the spread of pathogens in intensive farming systems, antimicrobials such as avoparcin and virginiamycin are used to promote growth in pigs, chickens and feedlot cattle.
- Pesticide resistance
  - Insecticides, acaricides, herbicides and anthelmintics are chemicals used to manage macroparasites and weeds on farms. Their overuse has led to the emergence of resistant forms of parasites and weeds, making it harder to manage infectious diseases on farms.
- Loss of genetic diversity
  - Genetic variation is necessary for a population to evolve in response to a disease threat. The use of **inbreeding** in animals and plants, or **monoculture** practices in plants, can lead to reduced resilience of a population to a new pathogenic threat.
- Increase in 'hobby farmers'
  - As Australian urban populations swell, many people seek an alternative life style or 'tree change'. People with little knowledge or experience of **animal husbandry** may be unaware of the risks of certain practices.

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- Increase in use of aquaculture as marine and freshwater animal populations decrease
  - Aquaculture involves the farming of seafood. It is a growing export industry in Australia as the world population increases and the need for a source of protein increases, but wild stocks decrease due to overfishing, habitat loss and marine pollution. Species include fish such as salmon, tuna and barramundi, oysters, abalone, crab, prawns and lobsters.
  - Antimicrobials are used to **therapeutically** and **prophylactically** control the outbreak of bacterial and fungal diseases due to the close proximity of organisms and the increased risk of **cross-infection**. Common bacterial threats include *Aeromonas* spp., *Pseudomonas* spp. and *Vibrio* spp. No antibiotics are currently registered for use in aquaculture but there may be pressure to use antibiotics off-label.

Plant diseases of agricultural significance

A range of plants are grown commercially in Australia:

- grains – the seed of a cereal crop, such as wheat, corn and barley, for human and animal consumption
- fruits and vegetables – for domestic consumption and export
- fodder – food for livestock, such as oaten hay, alfalfa hay and silage
- fibre – filaments or threads from plant material used for textiles, such as cotton
- horticultural plants – plants cultivated for use in gardens and orchards
- forestry plants – plants used in the creation and conservation of forests for human and environmental benefits.

Australia is relatively free of most of the world's most damaging plant pest species. However, there is still an array of pathogens affecting Australian plants that are a threat, not only to the natural environment but also to our agricultural industries.

Causes of infectious diseases in plants in agriculture

In the natural environment, plants are commonly attacked by pathogens. Plants have evolved unique ways to keep these pathogens in check, such as the **abscission** (dropping) of infected fruits or leaves. When plants are grown for horticultural or agricultural purposes, they are grown in higher densities, and pathogens that were not considered a problem previously may become significant. If soil pH, nutrient balance and water availability are not optimal, the stress caused to plants can reduce their natural ability to inhibit pathogen invasion and growth. Many of these pathogens gain entry through natural openings in the plants, such as stomates, or wounds caused by insect bites or other damage due to hail, wind or pruning.

The majority of plant infectious diseases are caused by the types of pathogens described below.

Fungi

Fungi are by far the most common cause of plant disease. Terms such as 'rust', 'smut', 'blight' and 'mildew' are used to describe fungal diseases in plants. Some have colourful names such as *gummy stem blight* and *white blister* (Fig. 10.44). Reservoirs of fungal spores exist in contaminated seeds, farm machinery, soil and nearby weeds, and are generally transmitted by wind, water and contact with the reservoirs through normal farming operations. Fungi enter plants through their stomata or any other opening caused by mechanical damage to the plant, such as pruning and insect bites. They damage the plant by destroying conducting tissues and absorbing nutrients from the plants.

FIGURE 10.44 White blister disease looks like large white blisters on the leaves.

FIGURE 10.45 The characteristic tunnels made by the citrus leaf miner

FIGURE 10.46 Bacterial disease causes the tissues of the plant to rot, change colour and become slimy

FIGURE 10.47 Nematode eggs are clearly visible on these soybean roots.

FIGURE 10.48 The mottled appearance of a tomato leaf infected with tomato virus

Insects and mites

Insects and mites not only cause direct damage to plant tissue, but may also act as vectors for other pathogens. Examples are aphids, fruit fly, citrus leaf miner and mealybugs. The citrus leaf miner is a moth that lays its eggs under the leaf of the citrus plant. The larvae hatch and burrow through leaves, leaving characteristic 'mines' (Fig. 10.45), and the leaves then twist and curl. Young plants are most at risk as their growth can be severely inhibited.

Bacteria

Reservoirs of pathogenic bacteria may occur in soil, weeds and seeds. Humans can also harbour bacteria on their hands and equipment from previous work with a contaminated crop of plants. However, bacteria only multiply and spread when certain conditions are met. These include humid, warm weather, overcrowding of plants, inappropriate soil conditions (water, nutrients, pH and salinity) and poor air circulation. Examples of bacterial diseases (Fig. 10.46) include black rot of brassicas, bacterial canker of tomatoes and bacterial blight of peas. *Pseudomonas spp.* are particularly common bacterial pathogens of plants as they are capable of tolerating a wide range of conditions.

Nematodes

Thousands of nematode species live in soil but only a few act as plant pathogens. An example is the root knot nematode (Fig. 10.47), a pathogen of agricultural significance, particularly for tomato growers. The nematode attacks plant roots, creating galls and lumps. The plants subsequently wilt, turn yellow and die. The eggs of these nematodes can persist in the soil for a year and reinfect the next crop. The infestation can be dealt with by repeated cultivation of the soil and exposure to the sun, combined with removal of residual root material after harvesting to reduce reservoirs of the eggs.

Viruses

Plant viruses are obligate intracellular parasites and are less well understood than animal viruses. The first to be discovered was the tobacco mosaic virus, which infects tobacco plants and produces a mottling pattern on the leaves. The tomato mosaic virus and the pepper mild mottle virus are other examples (Fig. 10.48). All these viruses are stable in the environment and can persist in plant material left over after cropping. They can also form a reservoir on contaminated equipment. Increased plant densities and frequent handling of plants by humans appear to play a role in its transmission.

Phytoplasmas

Phytoplasmas are related to bacteria but do not have a cell wall. They are transmitted from plant to plant by insect vectors, and inhabit phloem tissue. They are pathogens of agricultural importance because they have been reported in plants such as tomatoes, strawberries, grapes and pumpkins (Fig. 10.49).

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Case study: Panama disease of bananas

Bananas are the largest horticultural industry in Australia and a best-selling product in our supermarkets. Most banana plantations are in North Queensland, which produced 95% of Australian bananas in 2014–15. In March 2015, the pathogen that causes Panama disease of bananas was detected on a property that grew Cavendish bananas in the Tully Valley, North Queensland. Cavendish bananas are the main type grown in Australia. Panama Tropical Race 4 disease is caused by the highly contagious fungus *Fusarium oxysporum*. It causes yellowing and wilting of leaves and splitting of stems (Fig. 10.50). The conducting tissues are damaged and so the plant is starved of water and food. The disease is spread through root-to-root contact and contaminated soil from machinery and shoes.

FIGURE 10.49 Symptoms of phytoplasma infection vary; they include yellowing, stunting and 'witches' brooms (many small, distorted shoots growing clumped together).

Farming operations ceased and the affected property was sold to the Australian Banana Growers Council, because the fungus contaminates the soil permanently, and remains a biosecurity risk. No other plants can be grown commercially on this property for this reason. The perimeter fences were reinforced, all banana plants were destroyed and stabilising ground cover was established to prevent soil runoff into neighbouring properties and waterways. Strict quarantine rules enforced by Biosecurity Queensland have contained the outbreak to one farm so far. During the outbreak, the price of bananas skyrocketed in Australian supermarkets.

FIGURE 10.50 Stem of a banana plant with Panama disease. This cutaway of the stem shows the most characteristic disease symptom–the reddish-brown discolouration of the water-conducting tissues

Abiotic factors that cause disease

At the start of this chapter, you read that disease arises from an imbalance between the pathogenicity of the agent and the defences of the host. Abiotic factors play a major role in setting a plant up for invasion by a pathogen. These factors include any major alterations in:

- temperature variation
- light availability
- chemical agents (natural and synthetic)
- water quantity and quality
- nutrient availability in soils.

As with infection of animals, for infectious diseases of plants to exist, there must also be a chain of infection.

Effects of infectious diseases in plants

The effects of infectious disease in plants can be considered at three levels:

- biological effects on the individual plant
- social and economic effects on the farmer
- social and economic effects on Australia's economy.

These effects are discussed on the following page.

Weblink
Panama disease

Biological effects of pathogens on individual plants

Some of the ways in which pathogens disrupt the normal operation of plant tissues are summarised in Table 10.7. These symptoms often occur in combination.

TABLE 10.7 Effects of pathogens on plant tissue

SYMPTOM	FEATURES	SYMPTOM	FEATURES
Death of plant	Plants lose their ability to balance water uptake with water loss through diseased conducting tissue, or the ability to produce food (photosynthesise) because of loss of photosynthetic tissue.  **FIGURE 10.51** Jarrah forest infected with 'dieback' fungus, *Phytophthora cinnamomi*	Destruction of tissues (necrosis)	Cell death can be caused directly through pathogen attachment and invasion of cells or indirectly though the effects on photosynthetic and conducting tissues.  **FIGURE 10.52** Spots of necrosis on the leaves of a hazelnut tree, caused by the bacterium *Xanthomonas arboricola*
Abnormal growth	Normal plant growth is regulated by a series of complex interactions between hormones (trophic factors) and cells. Disease processes may interfere with the production, distribution and action of hormones. Tumour-like galls are a common sign of infectious disease.   **FIGURE 10.53** Gall wasp infestation in a citrus tree: **a** wasp larvae emerging from a gall; **b** wasps secrete growth-regulating chemicals that cause abnormal swellings (galls) to develop.	Discolouration of tissues	Leaves may turn yellow (chlorosis), indicating a problem with the production of chlorophyll. Mosaic patterns are common with viral infections.  **FIGURE 10.54** Peanut leaf with concentric ring spots caused by peanut mottle virus
		Wilting	Wilting happens when the plant loses more water than it takes up from the soil. Causes include root damage, and damage to or interference with conducting tissues.  **FIGURE 10.55** Wilt in a tomato plant caused by the bacterium *Ralstonia solanacearum*

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INVESTIGATION 10.5

Secondary-source investigation of plant diseases and their transmission

AIM

Describe one plant disease for each of the following six types of pathogens discussed in this section: fungi, insects and mites, bacteria, nematodes, viruses, phytoplasms

RESULTS

Create a table to summarise the plant diseases. The following table is a suggestion.

PATHOGEN (TYPE AND SCIENTIFIC NAME OF ONE EXAMPLE)	DISEASE NAME AND TRANSMISSION	DESCRIPTION OF DISEASE	ADAPTATION OF PATHOGEN FOR ENTRY AND TRANSMISSION*

*Complete this column after reading Section 10.4.

Social and economic effects of diseases in plants

The threat of infectious disease places a great burden on primary producers. Constant vigilance is required for early detection and management. The most important consequences of a plant disease outbreak on a farm include:

- reduced yields of grains, pastures, fruits and vegetables
- loss of trading opportunities, both nationally and internationally
- economic loss for the farmer, resulting in financial hardship and stress for family and local community.

The Australian economy relies heavily on the export of grains, fruits and vegetables to overseas markets. Australia's physical isolation and the consequent disease-free status of much of its produce gives us unique access to markets around the world. The introduction of an exotic plant disease into Australia could have dire consequences for the national economy.

Animal diseases of agricultural significance

The international body that oversees and coordinates the management of animal diseases at a global level is the World Organisation for Animal Health (OIE, originally *Office International des Epizooties*). An **epizootic** is the animal equivalent of a human **epidemic**. As a member nation, Australia must report any exotic animal disease that it detects within its borders, especially those with a risk of transmission to humans (**zoonosis**). Infectious diseases of agricultural concern in Australia, including endemic and exotic diseases, are listed in Table 10.8.

Table 10.8 and the weblinks list a number of animal diseases that affect agriculture. You may wish to examine these further as a depth study.

Weblink
The Australian Veterinary Association
Detailed information regarding livestock diseases and their management

Weblink
World Organisation of Animal Health

Weblink
Animal disease information

Weblink
Biosecurity

Weblink
Farm biosecurity

Weblink
Aquaculture industry in Australia

TABLE 10.8 Some infectious diseases of agricultural concern in Australia

PATHOGEN TYPE	EXAMPLES OF INFECTIOUS DISEASES*
Bacterium	• Mastitis • Foot rot • Q fever • Brucellosis • Leptospirosis • Clostridial diseases such as black disease, blackleg, tetanus, pulpy kidney, malignant oedema • Tuberculosis • Pink eye
Virus	• Three-day sickness (bovine ephemeral fever) • Foot and mouth disease • RHDV1 (calicivirus) in rabbits • Rinderpest • Bluetongue • Rabies • Coronavirus • Avian influenza • Swine flu • Avian and porcine circoviruses
Fungus	• Lumpy jaw (actinomycosis) in cattle • Aspergillosis • Ringworm (dermatophytosis) • Cryptococcosis • Blastomycosis
Protozoan	• Coccidiosis • Toxoplasmosis • Cryptosporidium • Babesiosis • Trypanosomiasis • Leishmaniasis
Prion	• Scrapie in sheep • Bovine spongiform encephalopathy • Chronic wasting disease of cervids (deer, moose etc)
Macroparasite	• Liver fluke • Roundworm • Flystrike

*Including endemic diseases and exotic diseases not currently found in Australia

INVESTIGATION 10.6

Secondary-source activity on diseases of agricultural significance in Australia

Table 10.8 lists some of the major diseases of agricultural significance in animals. Some are found in Australia but many are exotic diseases not currently found in Australian agriculture.

AIM

To choose an infectious disease of agricultural significance and develop an advertisement that could be used to convince farmers to change their practice to prevent the spread of the named plant or animal disease

COMMUNICATION

Choose a suitable medium for the advertisement. Peer evaluation of your argument must be completed prior to submission. Your advertisement must use suitable scientific terminology and provide accurate advice to farmers. »

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» METHOD

1 Choose one of the diseases listed in Table 10.8, *or* select a disease that you know of. Discuss this with your teacher first. Choosing an exotic disease is fine, because it could pose a serious threat to Australian agriculture, should there be an outbreak here.

2 In groups of two or three, use secondary sources such as printed and online materials to conduct the following research:

   a Identify the pathogen (common and species name).

   b Name the disease. Many of these diseases have a scientific as well as a common name. For example, bovine spongiform encephalopathy is also known as 'mad cow disease'. Try to identify as many common names as possible for your chosen disease.

   c Outline the signs/symptoms of the disease.

   d How is this pathogen transferred? What are the conditions on a farm that favour transmission of the pathogen? Think about the three factors that influence infectious disease outbreaks (host, pathogen and environment).

   e Read ahead in this chapter (Section 10.4) and identify some of the possible pathogen factors that favour the transmission and disease-causing ability of your chosen pathogen.

   f Identify the treatment for this disease. Sometimes this will involve management on a whole-farm scale, not just treatment of an individual animal. 'Herd health' refers to a wholistic approach to disease on a farm, concentrating on all factors at once.

   g Identify some online resources that farmers can refer to, for advice on this disease.

   h Find out whether the disease is **notifiable**. 'Notifiable' means that by law the disease must be reported by a veterinary surgeon to a government agency, for reasons of national biosecurity. For example, foot-and-mouth disease is notifiable throughout Australia.

RESULTS

Select the type of presentation that is most suitable. This may be a simple table, or perhaps a more visual form of presentation such as a slideshow.

DISCUSSION

In what ways can a manager of an agricultural enterprise ensure that this particular pathogen does not become a problem?

CONCLUSION

What are some general recommendations your group could make to a farmer regarding this disease?

Effects of infectious diseases in farm animals

Different diseases in farm animals can affect primary producers in different ways, including:

- death of the affected animals (e.g. Black's disease, anthrax)
- loss of appetite and weight over a short or extended period (e.g. three-day sickness in cattle)
- economic loss to the farmer, with negative effects on profitability and production due to reduced meat, milk and wool yields
- loss of international trading opportunities if Australia's disease-free status is threatened (e.g. if foot-and-mouth disease became endemic in Australia)
- human illness and disease (zoonoses) (e.g. Q-fever, brucellosis, leptospirosis)
- low growth rates in young animals (e.g. internal parasites such as worms)
- loss of fertility in females through embryonic death or stillbirths (e.g. leptospirosis, brucellosis)
- loss of economic value of individual animals due to blemishes or ectoparasites (e.g. warts in beef cattle).

KEY CONCEPTS

- Australia relies heavily on its agricultural exports, because of its unique geographic isolation and the disease-free status of much of its produce.
- Both endemic and exotic pathogens pose a risk to Australian agriculture.
- Disease outbreaks on farms result from the interplay of host factors, pathogen factors and environmental factors.
- The risk of infectious disease is a growing threat, due to at least nine major factors including: increased human mobility, the rise of industrial agriculture, changing patterns of land use, climate change, and antimicrobial and pesticide resistance.
- Animal and plant diseases can have dire consequences at the farm level and at the national level, and must be well managed.
- Local, national and international bodies exist to manage biosecurity issues arising from plant and animal diseases.

CHECK YOUR UNDERSTANDING 10.3

1. What is the significance of Australia's relative geographical isolation for infectious diseases in plants and animals?
2. Distinguish between 'endemic' and 'exotic' diseases.
3. Describe three factors that contribute to an outbreak of infectious disease in an agricultural enterprise. Use an example of a named animal or plant disease to demonstrate this.
4. Discuss the factors that contribute to the increased risk of infectious diseases in Australian animals and plants.
5. Identify the most common entry point for most pathogens into plant tissues.
6. If you were working at a local garden centre, what signs of plant ill health would you need to be aware of?
7. Justify the formation of national and international organisations to coordinate the response to disease outbreaks of agricultural concern. Use at least two named examples.
8. List three economic impacts of infectious disease on the agricultural industry, at the level of individual farmer, the district they live in and the nation as a whole.
9. Match each term below with the correct figure.

Wilting	 Genevieve Vallee/Alamy Stock Photo FIGURE 10.56	Necrosis	 Daniel Allan/Getty Images FIGURE 10.58
Discoloration	 saiko3p/Shutterstock.com FIGURE 10.57	Abnormal growth	 evan66/Shutterstock.com FIGURE 10.59

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10.4 Adaptations of pathogens to facilitate their transfer

Many Hollywood blockbusters depict a hero or villain using some novel technique to get into a building (Fig. 10.60a). Their success or failure depends on their ability to adhere to the side of the building, evade detection and all the security devices in place, gain entry, and remain undetected or fight their way deeper inside.

In the same way, for a pathogen to successfully establish an infection, it must find a way to adhere to the host's cells, colonise the host tissues, spread to other tissues, and persist in the host long enough to reproduce. In other words, for an organism to cause disease, it must:

1. enter the host
2. multiply in host tissues
3. resist or not stimulate host defence mechanisms
4. damage the host.

As in the movies, adhesion precedes invasion (Fig. 10.60b).

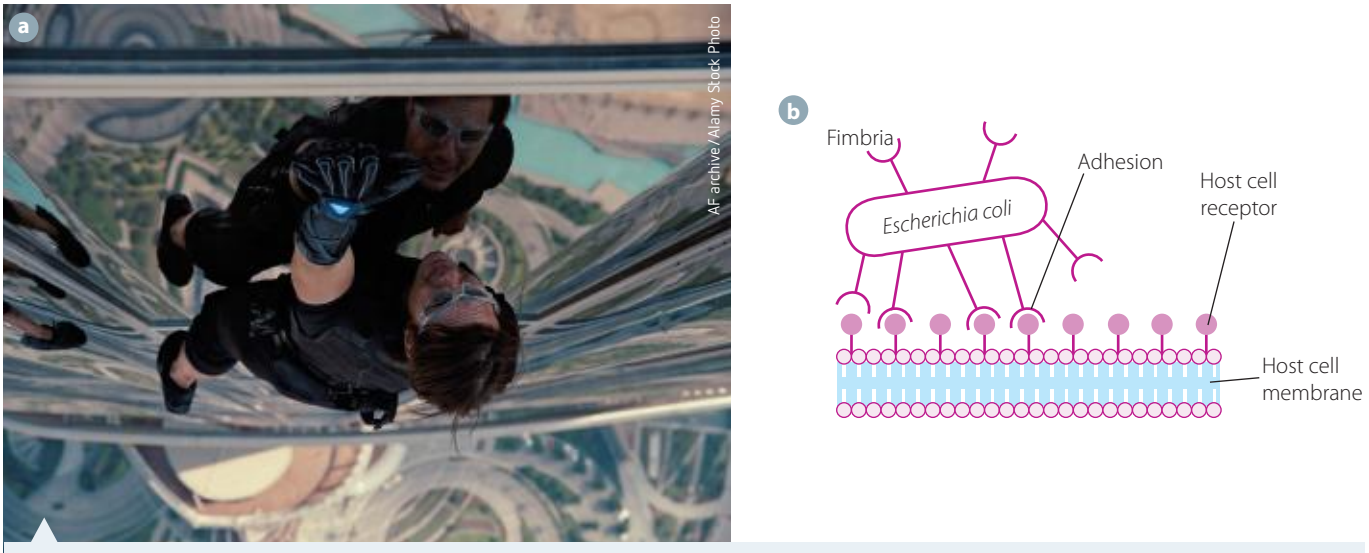
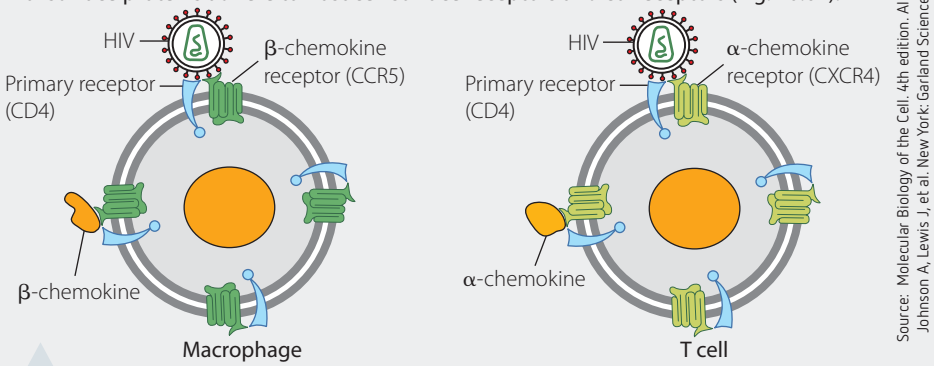
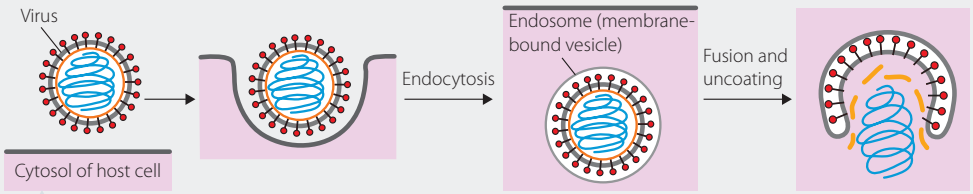


FIGURE 10.60 Adhesion and invasion: **a** Successful entry to a secure building involves good adhesion followed by invasion. **b** Bacteria adhere to a host cell using fimbriae (or pili, which are similar to fimbriae but longer).

Pathogens have developed an array of strategies or **adaptations** to enable them to adhere to, gain entry into and persist in their hosts. These strategies form part of the virulence factors for that pathogen. Each pathogen has a 'toolkit' of virulence factors that help it to successfully establish itself in host tissues. It is thought that the evolutionary strategies of pathogens are just slightly ahead of the development of host resistance strategies. The two have evolved more or less side by side throughout the history of life.

Table 10.9 summarises some of the strategies that the various cellular and non-cellular pathogens use to gain access to and colonise host tissues.

TABLE 10.9 Adaptations to facilitate adhesion to and invasion of host by pathogens

PATHOGEN	VIRULENCE FACTORS*
Prions	• Host B lymphocytes are thought to play a role by secreting factors (e.g. tumour necrosis factor) that enable prions to invade follicular dendritic cells in lymphoid tissue. • From lymphoid tissue, they invade nervous tissue through the autonomic nerves and travel to the brain. • May 'piggyback' other proteins such as ferritin (abundant in meat) to facilitate movement through the gut.
Viruses	Adhesion: • Must enter the nucleus of the host cell to facilitate replication of the viral genome. • Viral surface proteins adhere to host cell surface receptors and co-receptors (Fig. 10.61).  Source: Molecular Biology of the Cell. 4th edition. Alberts B, Johnson A, Lewis J, et al. New York: Garland Science; 2002. **FIGURE 10.61** Viral adhesion: HIV virus adhering to a host macrophage and T cell via receptors on the surface of the cell Source: Molecular Biology of the Cell. 4th edition. Alberts B, Johnson A, Lewis J, et al. New York: Garland Science; 2002. Invasion: • Receptor-mediated endocytosis (movement of the virus into the cell). Enveloped viruses (e.g. influenza) are enclosed within an envelope (endosome) formed from the host cell membrane as they move into the cell (Fig. 10.62). • Non-enveloped viruses (e.g. polio virus) form a pore in the host cell membrane and deliver the viral genome through it. • Some viruses use the cell's normal membrane-forming processes, follow a route through the endoplasmic reticulum and Golgi body and then bud off from the surface.  **FIGURE 10.62** Receptor-mediated endocytosis of an influenza virus
Bacteria	Adhesion: • Pili and fimbria • Adhesins on the surface of the bacterial cell resist washing action of secretions such as urine, mucus, cilia. • Translocation of bacterial proteins cause host cell membrane engulfment of bacteria. • Bacterial cells form a biofilm. Invasion: • Enzymes such as collagenase, hyaluronidase and lecithinase break down cell contents. • Capsules resist phagocytosis by host cells. • Intracellular bacteria (e.g. tuberculosis): phagocytosis by macrophages and walling off in granulomas (tubercles). • Chemical strategies to destroy host immune defences, such as leucocidin, IgA protease. • Host cell cytoskeleton is used for intracellular movement. • Toxins are secreted to damage host cells (endotoxins and exotoxins). • If phagocytosed, *Listeria monocytogenes* secretes haemolysin, which selectively destroys phagosomal membrane but not cell membrane.
Protozoan	• *Toxoplasma gondii* (toxoplasmosis): microtubule protrusion into host cell facilitates entry (intracellular parasite), and formation of a vacuolar membrane gives protection from lysosomes. • *Trypanosoma cruzi* (Chagas disease) – in receptor-mediated attachment, recruits lysosomes to fuse with cell membrane. Pathogen enters vacuole made of lysosomal membrane, then deactivates lysosomal enzymes.

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PATHOGEN	VIRULENCE FACTORS*
Fungus	Adhesion: • Assisted by cell wall or capsule molecules that permit adhesion to host cells. Invasion: • Thermotolerance – heat shock proteins are synthesised to cope with body temperatures (higher than air temperature). • Converts from saprophytic mycelium to parasitic yeast when exposed to heat (dimorphism) (Fig. 10.63). • Cell wall and capsules protect fungi from host attacks, e.g. alpha glucan is a cell wall polysaccharide that confers protection. • Hormone receptors for 17B-oestradiol on fungal cells may change the incidence of certain fungal diseases between men and women. • Secretion of hydrolytic enzymes causes damage to host cells and provides nutrients for fungus. • Evasion mechanisms include capsule production, suppression of cytokine production by host cells, and reduced fungicidal power of macrophages. • Opportunistic fungal infections (e.g. *Cryptococcus neoformans*) are common in immunosuppressed patients (e.g. HIV/AIDS, chemotherapy, lymphoma)
Macroparasites	• Tapeworms (*Taenia* sp.)- attach themselves to the inside of the intestine of their host using their scholex (head), which may have either hooks, suckers or both (Figure 10.64). They do not have a mouth but instead absorb nutrients directly from the host's digestive tract via their tegument, which both protects the parasite from digestive enzymes and allows for gas exchange to occur. • Ticks – highly specialised mouthparts are inserted into host skin to attach. Tick is anchored in skin by 'attachment cement'. Biologically active molecules are secreted in saliva to prevent vasoconstriction, and prevent host from forming a clot or initiating an inflammatory response.

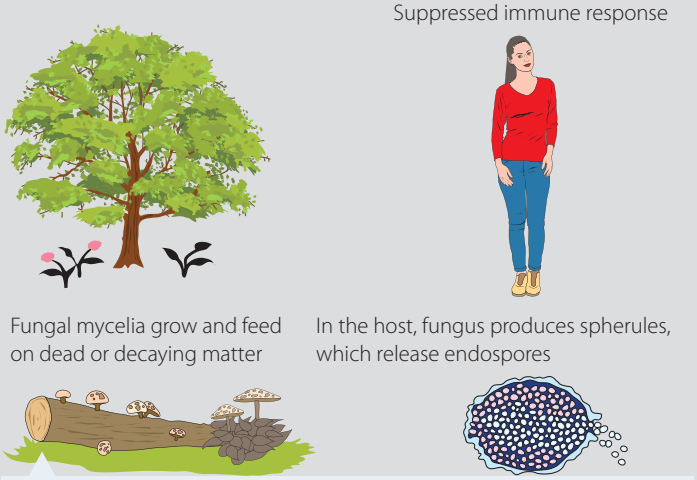
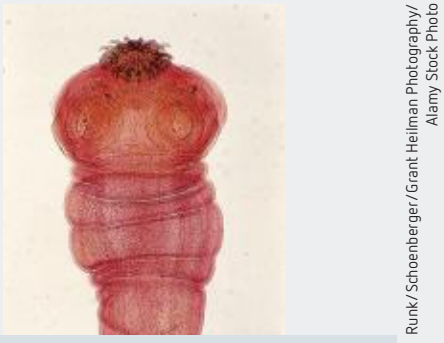
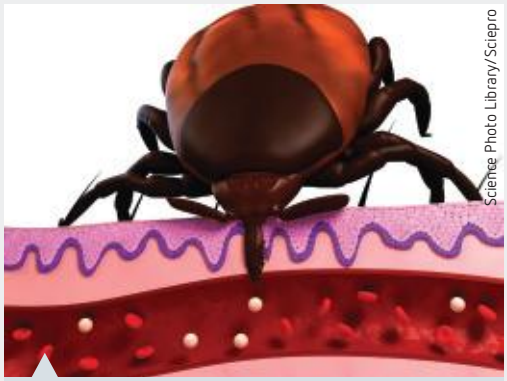


FIGURE 10.63 Factors that allow fungal invasion of a host



Runk/Schoenberger/Grant Heilman Photography/Alamy Stock Photo

FIGURE 10.64 Micrograph of the scolex (head) of *Taenia solium*, showing the hooks and suckers used for attachment to the host's gut.



Science Photo Library/Scienpro

FIGURE 10.65 The specialised mouthparts of a tick facilitate the transfer of pathogens.

*Adaptations to facilitate adhesion to and invasion of host

Adaptations to facilitate transmission between hosts

Transmission of a pathogen refers to the passing of the pathogen from one host to another, directly or indirectly:

- direct transmission through sneezing, coughing, sexual contact
- indirect transmission by touching a contaminated surface or object or via a vector.

Table 10.10 summarises some of the adaptations acquired by pathogens to exploit their own unique method of transmission.

TABLE 10.10 Adaptations to facilitate transmission of pathogens

TRANSMISSION ROUTE	ADAPTATIONS TO FACILITATE TRANSMISSION	EXAMPLES OF PATHOGENS
Airborne on dust and respiratory secretions (e.g. water droplets)	• Able to remain suspended in air for long periods. • Resists drying out. • Pathogen causes sneezing and coughing, which causes ejection and transmission to new host. • Aero-tolerant – able to tolerate a wide range of oxygen concentrations.	• influenza viruses
Waterborne	• Able to colonise and proliferate in water, so environmental reservoir is present (e.g. from faecal material). • Modified outer surface structures (e.g. fimbria, flagella) allow motility. • Marine-borne organisms are halotolerant (able to tolerate high salinity). • Many are not destroyed by simple boiling of water or other water-treatment processes.	• *Legionella* • *Vibrio* • *Giardia* • *Campylobacter* spp.
Vector-borne	• Poorly understood at present. • Changing type and number of vectors used as reservoirs and for transmission. • Vector is not affected by pathogen. • Form preferential reservoirs in digestive tract or salivary glands of vector for easier transmission. • Produce special surface proteins that allow attachment to vector tissues. • Life cycle of pathogen is synchronised with feeding habits of hosts.	• *Rickettsia felis* now uses fleas and mosquitoes as vectors • malaria • Zika virus • Hendra and lyssa viruses
Faeco-oral (Fig. 10.66)	• Pathogens are generally very stable in varied environments, e.g. acid in stomach, low oxygen of large intestine. • Induction of vomiting and diarrhoea increases likelihood of transmission. • Antimicrobial resistance genes.	• *E. coli* • *Salmonella* spp.
Soil-borne	• Form endospores to resist desiccation. • Stable in the environment under a range of conditions. • Grow mainly in the root zone (rhizosphere). • Only a few bacteria are soil-borne pathogens of plants.	• *Clostridium tetani* • fungi • nematodes

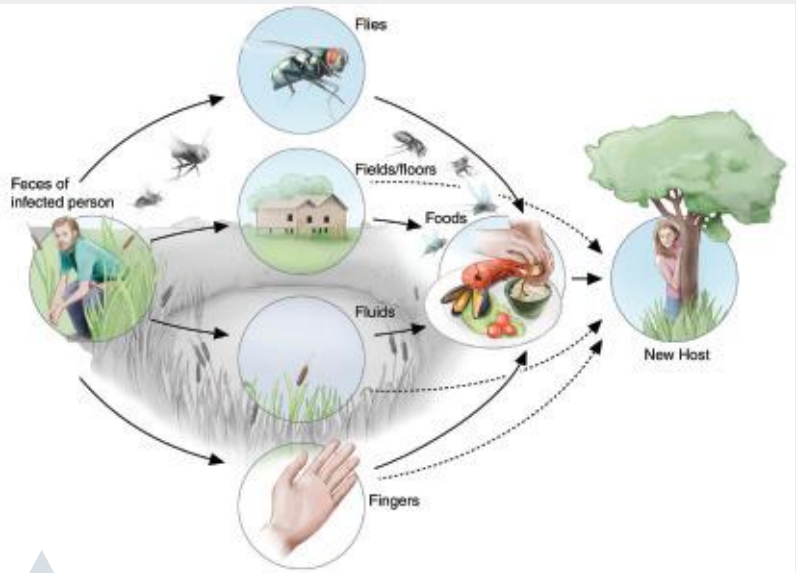


FIGURE 10.66 Faeco-oral transmission of pathogens

Source: deGraaf M, Beck R, Caccio S et al. Sustained fecal-oral human-to-human transmission following a zoonotic event, Current Opinion in Virology, Vol 22, Figure 1, pp. 1–6. Feb. 2017, With permission from Elsevier.

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TRANSMISSION ROUTE	ADAPTATIONS TO FACILITATE TRANSMISSION	EXAMPLES OF PATHOGENS
Sexual (venereal)	• Same as for vertical (See below.)	• chlamydia • HIV/AIDS virus • gonorrhea • herpes simplex virus
Blood-borne	• Take advantage of altered features of red cells to facilitate growth and development.	• malaria parasites and sickle cell anaemia
Vertical (mother to child)	• Capable of transmission across the placenta where maternal and foetal cells juxtapose. • Capable of uterine invasion. • Unprotected sexual activity facilitates transmission. • Consumption of placenta by other animals in the wild facilitates transmission. • May be aerosolised from afterbirth.	• *Brucella* spp. (contagious abortion in cattle) • *Parvovirus* • rubella virus • chickenpox virus • *Listeria monocytogenes* • *Plasmodium falciparum*

KEY CONCEPTS

- Virulence factors help a pathogen to be transmitted and gain entry to a new host.
- For infection to occur, the pathogen must be transferred to the host and then adhere to and invade the host.
- The transfer of a pathogen between hosts may be direct or indirect.
- Pathogens have traits that maximise the success of their transfer, adhesion and invasion.

CHECK YOUR UNDERSTANDING 10.4

1 Define the following terms.
   - **a** adhesion
   - **b** fimbria
   - **c** pili
   - **d** virulence factors
   - **e** endocytosis
   - **f** invasion

2 Identify the four steps necessary for a pathogen to cause disease.

3 Describe one adaptation of each of the following pathogens that facilitates adhesion to or invasion into host cells.
   - **a** prions
   - **b** viruses
   - **c** bacteria
   - **d** protozoa
   - **e** fungi
   - **f** macroparasites

4 What does 'transmission' mean in reference to the spread of infectious diseases?

5 Create a table to compare the adaptations of different types of pathogens for entry into and transmission between hosts.

6 Describe an important strategy of a water-borne organism that facilitates transmission.

7 Where do vector-borne pathogens form preferential reservoirs? What do you think is the significance of this for disease transmission?

8 Identify a water-borne pathogen and outline a strategy it uses to facilitate transmission.

9 Justify the statement that no one type of pathogen is any more dangerous than another.

10 How could a newborn baby pick up an infectious disease from the mother?

10 CHAPTER SUMMARY

Cause and transmission of infectious disease: How are diseases transmitted?

WHAT IS INFECTIOUS DISEASE?

Disease is any process or condition that adversely affects the normal functioning of a living thing or parts of a living thing. An *infectious* disease is caused by (caught from) another organism or infective *agent* known as a *pathogen*.

EXAMPLES OF DISEASES CAUSED BY PATHOGENS

diseases caused by bacterial pathogens

BACTERIA	NAME OF DISEASE	FEATURES OF DISEASE
Bordetella pertussis	Whooping cough (pertussis)	• Runny nose, sneezing • Characteristic 'whoop' during coughing bouts • Gagging or vomiting
Salmonella enterica	Salmonellosis (food poisoning)	• Vomiting and diarrhoea • Dehydration • Fever and abdominal cramps

diseases caused by fungal pathogens (mycoses)

FUNGUS	NAME OF DISEASE	FEATURES OF DISEASE
• *Epidermophyton* • *Trychophyton* • *Microsporum*	• Tinea • Tinea • Ringworm	• Cutaneous mycoses • Fungus secretes enzymes that break down keratin • Exposure to pathogen combined with warm, moist conditions necessary • Redness, itching, scaliness of skin • Ringworm may cause classic ring-shaped lesions on skin • Yellowing and hardening of nails

diseases caused by protozoal pathogens

PROTOZOAN	NAME OF DISEASE	FEATURES OF DISEASE
Plasmodium spp.	Malaria	• Fever • Fatigue • Headaches • Jaundice • Vomiting

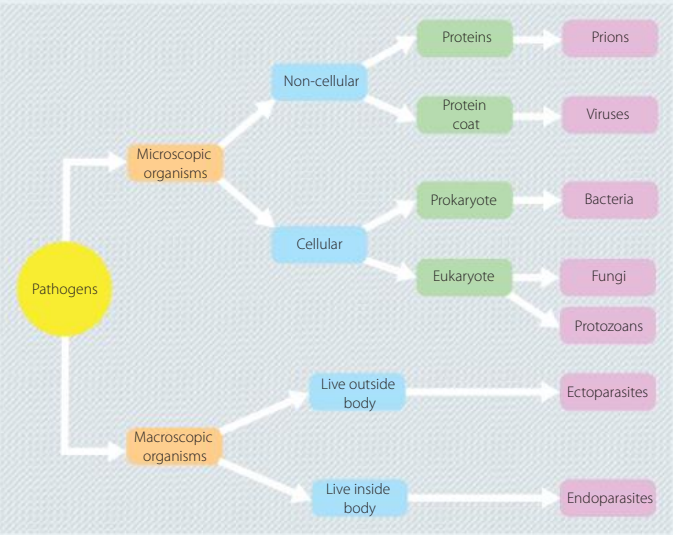
helminth diseases in animals

HELMINTH	NAME OF DISEASE	FEATURES OF DISEASE
Enterobius vermicularis	Threadworm/pinworm (most common worm infection in Australia)	• Red and itchy anus • Irritability and behaviour changes • Lack of appetite • Restless sleep • Worms may be visible in faeces • Weight loss • Abdominal pain • Diarrhoea • Blood in faeces
Taenia solium (pigs) *Taenia saginata* (cattle)	Tapeworm	
Toxocara canis (dogs) *Toxascaris leonina* (cats) *Ascaris lumbricoides* (humans)	Roundworm	

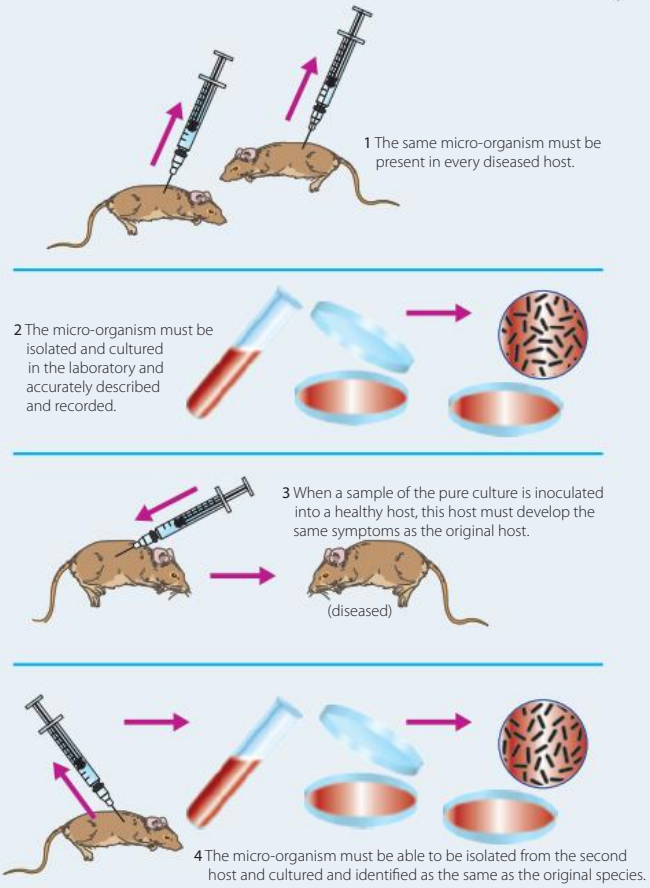
mites that infest animals and humans

MITE	DISEASE	FEATURES
Demodex canis (dogs)	Demodectic mange	• Overpopulation of the parasitic organism due to immune compromise of the host • Hair loss (alopecia) • Itching • Red skin • Crusting of skin

CLASSIFYING PATHOGENS



KOCH'S POSTULATES



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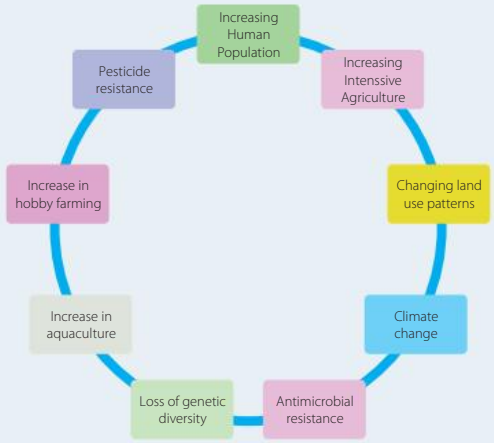
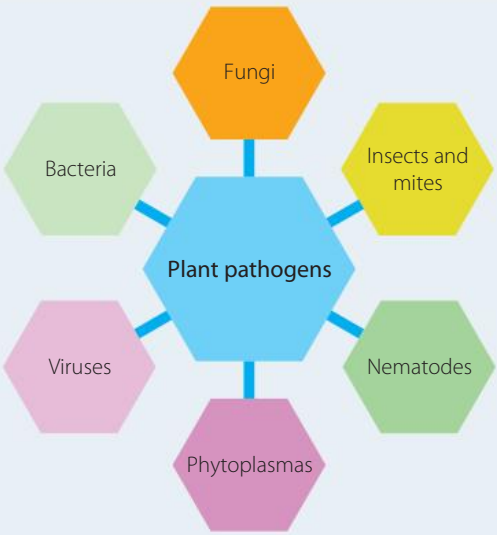
INFECTIOUS DISEASE AND AUSTRALIAN AGRICULTURE



Plant pathogens



TRANSMISSION MAY BE ASSISTED BY:

- secretion of chemicals (TNF in prions)
- surface receptors (viruses)
- pilli and fimbria (bacteria)
- microtubules (protozoa)
- adhesion molecules (fungus)
- immunomodulation (macroparasites)
- mouthparts (ticks).

To enter an individual host and cause disease, pathogens must:

1 enter the host
2 multiply in host tissues
3 resist or not stimulate host defence mechanisms
4 damage the host.

Effects of infectious diseases in farm animals

Different diseases in farm animals can affect primary producers in different ways, including:

- death of the affected animals (e.g. Black's disease, anthrax)
- loss of appetite and weight over a short or extended period (e.g. three-day sickness in cattle)
- economic loss to the farmer, with negative effects on profitability and production due to reduced meat, milk and wool yields
- loss of international trading opportunities if Australia's disease-free status is threatened (e.g. if foot-and-mouth disease became endemic in Australia)
- human illness and disease (zoonoses) (e.g. Q-fever, brucellosis, leptospirosis)
- low growth rates in young animals (e.g. internal parasites such as worms)
- loss of fertility in females through embryonic death or stillbirths (e.g. leptospirosis, brucellosis)
- loss of economic value of individual animals due to blemishes or ectoparasites (e.g. warts in beef cattle).

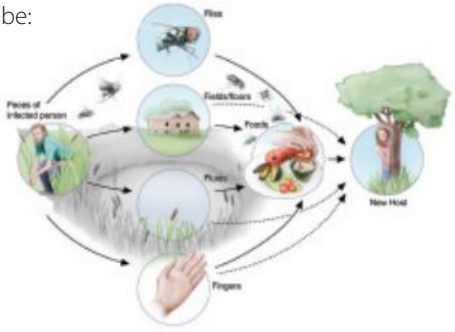
Effects of pathogens on plants

- death of plant
- destruction of tissues (necrosis)
- abnormal growth
- discolouration of tissues
- wilting

ADAPTATION OF PATHOGENS TO FACILITATE TRANSFER BETWEEN HOSTS

Transmission may be:

- airborne
- waterborne
- vector borne
- faeco-oral
- soil borne
- sexual
- blood borne
- vertical.



10 CHAPTER REVIEW QUESTIONS

Review quiz

1. Imagine a scenario where an outbreak of influenza is spreading rapidly through your school. What personal precautions would you take to ensure you did not become a host for the influenza virus?
2. Explain, using an example of an infectious disease, why the presence of a pathogen does not necessarily lead to the development of symptoms of an infectious disease.
3. Why do you think scientists are so obsessed with classifying organisms? What advantages could there be in classifying pathogens accurately?
4. Construct a table to compare and contrast the features of cellular pathogens. Carefully choose criteria that allow you to distinguish between them.
5. Design a hospital room in an infectious disease ward. Add features that you think would be useful to minimise transmission of pathogens from a carrier to doctors, nurses and visitors to the ward. Think about design features, movement of individuals, flow of materials such as wastes, air and materials that are easy to clean and launder.
6. Summarise the steps taken to control a particular infectious disease outbreak. Include an analysis of the effectiveness of these measures. Suggest possible improvements to the management of this disease outbreak.
7. Describe the method used to test for the presence of microbes in food and water. Justify *two* safety precautions taken during this investigation.
8. During your investigation of microbial testing of food and water, you researched the application of these techniques in the food industry. Use your knowledge of pathogens to justify *two* practices in the food service industry that are used to minimise pathogens in food or water.
9. Using a table, summarise the features of named diseases caused by direct, indirect and vector transmission.
10. Describe the contribution of Robert Koch to explaining the cause and transmission of infectious disease. In particular, justify the method he used to come to his conclusions.
11. Pasteur's experiments on microbial contamination showed definitively that the theory of spontaneous generation was false. Briefly outline the experimental method he used. Justify the design of his flasks. Explain in your own words why we can now be confident that pathogens must be transferred to a person for them to get an infectious disease.
12. Explain the possible effects on Australian agriculture of the introduction of new infectious plant and animal diseases.
13. Explain how a disease outbreak in a farm relies on *three* main factors. Use an example of an infectious disease in a farm animal.
14. Use a table to summarise the nine factors that increase the risk of infectious disease outbreaks in Australian agricultural enterprises.
15. Summarise the features of an infectious disease in a plant. What are the causes of this disease? What effect could it have on the individual farm and Australian agriculture as a whole (social and economic effects)?
16. Use a table to summarise the adaptations of pathogens to facilitate their transmission to new hosts.
17. How does the formation of endospores in certain bacteria facilitate their transmission to a new host? Use an example.
18. Outline the features of vertical transmission of pathogens. Use an example of a pathogen that transfers across the placenta.
19. Explain why the evolutionary adaptations of a pathogen need to stay a step ahead of those of their host organisms.
20. Describe methods used by pathogens to facilitate adhesion and entry into a host. Include both cellular and non-cellular pathogens.
21. Pathologists often take samples of infected tissue and attempt to culture the pathogen in broth or on agar. Use online sources to investigate the use of different culture mediums for bacteria and fungi.
22. Use an example to describe the social and economic effects of an exotic disease outbreak in Australia.
23. Justify the use of a control in an experiment you have completed during this chapter. Describe some ways in which you ensured that pathogens were excluded from this particular sample.

Exam preparation

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11 Responses to pathogens

INQUIRY QUESTION

How does a plant or animal respond to infection?

Students:

- investigate the response of a named Australian plant to a named pathogen through practical and/or secondary-sourced investigation, for example:
  - fungal pathogens
  - viral pathogens
- analyse responses to the presence of pathogens by assessing the physical and chemical changes that occur in the host animals' cells and tissues (ACSBL119, ACSBL120, ACSBL121, ACSBL122) CCT ICT

Luis Alvarez / DigitalVision / Getty Images

Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
11.1 Primary- and secondary source-investigation: Plant responses to pathogens
11.2 Secondary-source investigation: Christmas trees and asthma
11.3 Secondary-source investigation of urinary tract infections
11.4 The effect of temperature on the viability of pathogens

Worksheets
- Responses to pathogens
- First line of defence in the immune system
- Second line of defence
- Link between cytokines, inflammation and non-infectious diseases

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

FIGURE 11.1 Eucalypts killed by dieback, a fungal disease

> The states of health or disease are the expressions of the success or failure experienced by the organism in its efforts to respond adaptively to environmental challenges.
>
> – Rene Dubos, 1965
> Source: Dubos R. 'Man adapting'. New Haven: Yale University Press, 1965

When you walk through the Australian bush, you commonly see patches of dead eucalypts, sentinels of dieback, the water-borne disease caused by the fungus *Phytophthora cinnamomi* (Fig. 11.1). The fungus grows through the warm, damp soil and infects the roots of the tree, where the spores germinate and grow through the tree's root system. As it grows, the fungus releases water-borne spores, which swim through the water and infect healthy roots. The roots of an infected tree can no longer absorb the water required to sustain life, and the tree slowly dies of dehydration. The tree's immune response is triggered as the fungus invades the tissue of the plant root. The effectiveness of this immune response determines whether the plant lives or dies.

Worksheet
Responses to pathogens

This chapter examines the components of immune defence in plants and animals, and the role of each component in responding chemically or physically to the presence of a pathogen.

11.1 Investigating plant defences

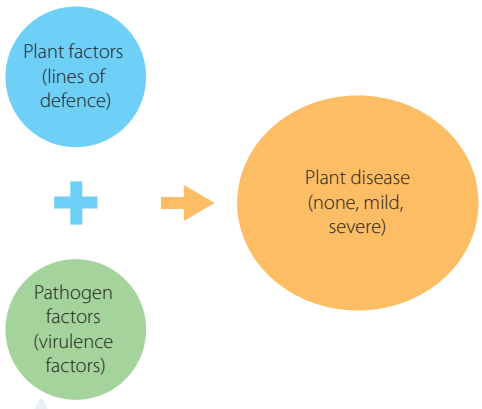

FIGURE 11.2 Infectious disease in plants depends on both host and pathogen factors.

When you look at a plant, from the outside it appears that not much is going on. However, plants are adept at avoiding infectious disease and responding to pathogenic threats. In nature, there are copious reservoirs of pathogens in the soil and water, and yet for the most part, plant populations survive and thrive, given the right conditions. However, plants grown as crops on farms may be more susceptible to pathogen invasion, as they are generally genetically uniform. We are all familiar with the responses of our own bodies when a cold or flu virus invades. We may have a fever, sore throat or muscle aches, all of which occur as part of the process of eliminating the pathogen. In animals we call the system responsible for this the **immune system**. But plants do not have an immune system in the way that animals do. They do not get a fever and shiver when they catch a virus. However, they do respond in observable ways to limit the growth or spread of the pathogen and save their own lives. And, just like us, they have a few well-developed lines of defence that help in this process.

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Plant responses to pathogens

In both natural and cultivated environments, plants have inherent disease-resistance strategies. These defences may be passive (such as physical and chemical barriers) or active (once the pathogen is recognised). If a plant can prevent a pathogen from invading its tissue, or prevent the pathogen from reproducing, then it will be resistant to that pathogen. This is determined in a complex interaction between the plant and the pathogen at the time of infection.

Passive defences

Plants have two major types of passive defences against pathogen invasion: physical barriers and chemical barriers.

Physical barriers

Physical barriers, such as a thick cuticle (Fig. 11.3), cell walls and small stomata, all inhibit pathogen entry. Some pathogens secrete enzymes to break down the cuticle, and so plants with thicker cuticles are better able to withstand this. Bark (Fig. 11.4) offers plants extra protection against pathogens that otherwise might invade and try to reach the food source, sap, in the phloem beneath the tree bark. Vertical hanging leaves, which do not accumulate a water film, reduce the likelihood of pathogen reservoirs building up on the outside of leaves. Stomata tend to open during humid weather and rainstorms, which helps regulate water balance in the plant, but is also a potential port of entry for pathogens.

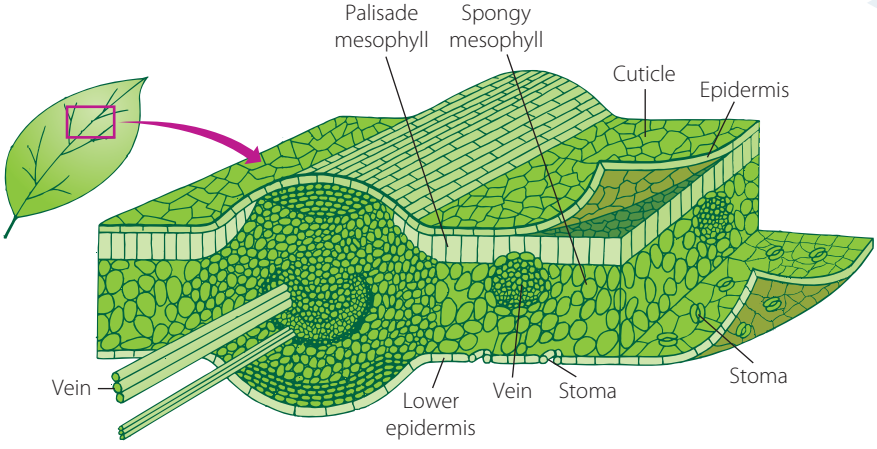


FIGURE 11.3 The cuticle and epidermis are a physical barrier preventing pathogen entry.

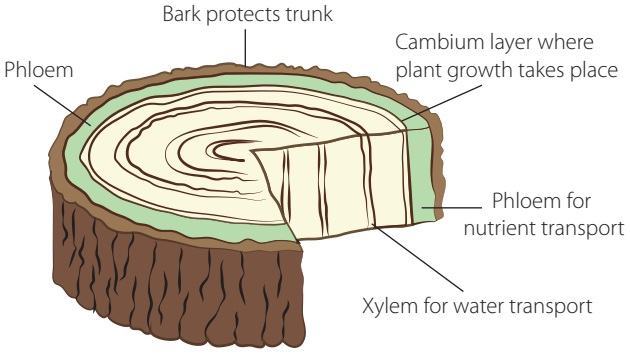
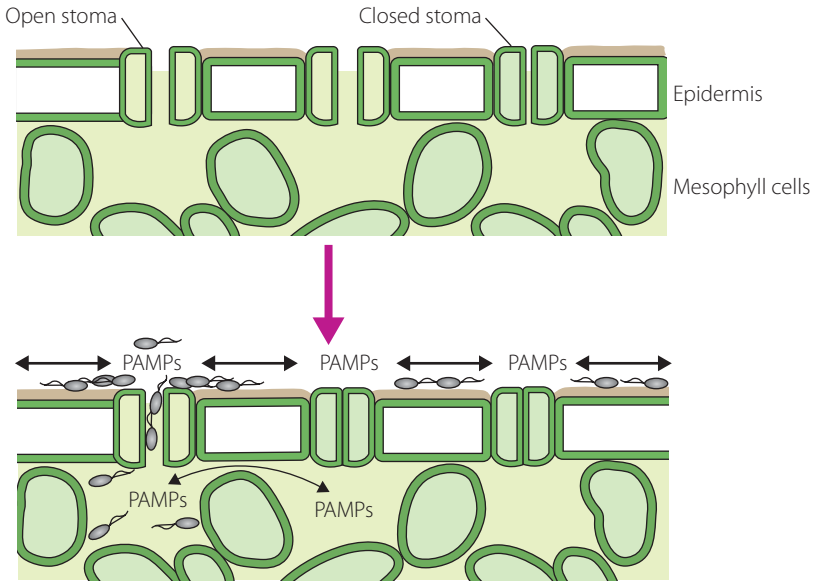


FIGURE 11.4 Bark is a strong physical barrier to pathogen entry.

Chemical barriers

Chemical barriers, such as the presence of chemical compounds in the tissues of plants, can reduce fungal and bacterial growth, and ward off vectors of viruses. Examples of chemicals are glucosides and saponins. Plants may also produce **enzymes** that break down pathogen-derived toxins. Chemical receptors on plant cells can detect the presence of a pathogen (for example, by detecting **pathogen-associated molecular patterns (PAMPs)** secreted by bacteria) and activate the next stage of defence (Fig. 11.5).

FIGURE 11.5
Stomata may close in response to the presence of bacteria, through chemical signalling methods involving detection of PAMPs.



Active defences

When its passive barriers are breached, the plant is now at grave risk of harm. The next line of defence involves more targeted responses by the plant. Three major groups of responses are involved: recognition of the pathogen, rapid response and delayed response.

Pathogen recognition

Plants are able to recognise pathogens by detecting certain physical and chemical signals, including fragments from the cell walls of bacteria and fungi. Genes within the cells of the plant are thought to regulate plant responses.

Rapid active response (minutes to hours)

Recognition of a pathogen by proteins on the surface of cells in plants causes changes in the permeability of the plant cell membrane. This allows the movement of certain ions (calcium ions in particular) into the cell and triggers defence responses by activating the expression of certain genes. The release of hydrogen peroxide (H_2O_2) in an **oxidative burst** can kill microbes directly. This release of hydrogen peroxide is often used in experiments as a chemical indication of a plant immune response. Another response is reinforcement of the cell wall with aggregates of material in the cytoplasm near a defect in the wall – this is known as *cell wall apposition* (Fig. 11.6). A third response is programmed cell death (**apoptosis**), which causes a cluster of dead plant cells to accumulate around the pathogen to isolate it, followed by the secretion of antimicrobial compounds.

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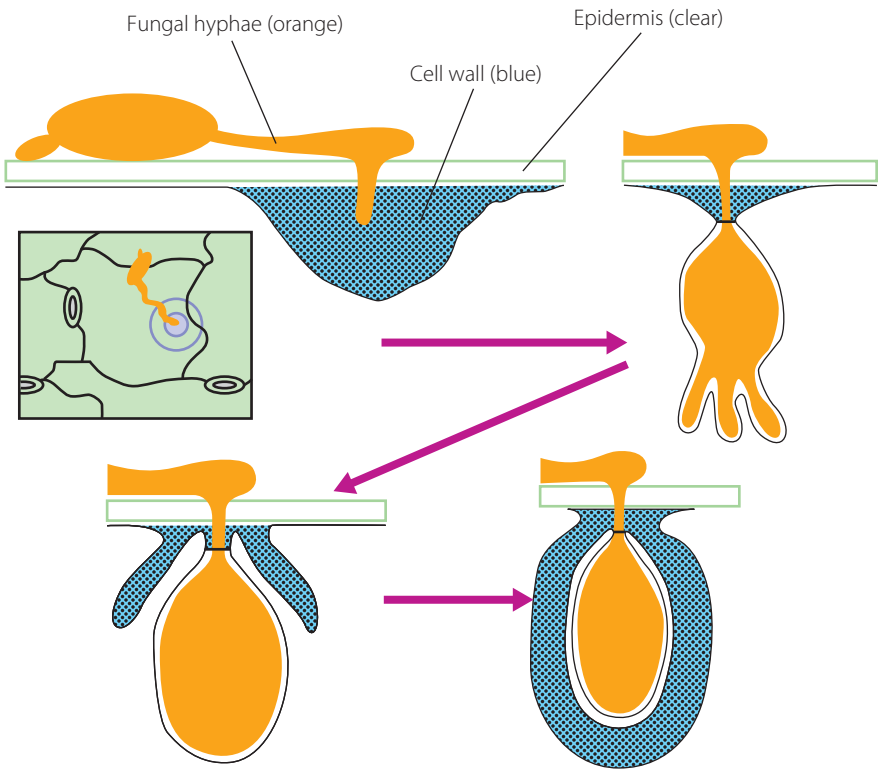

Source: Underwood W, The plant cell wall: a dynamic barrier against pathogen invasion. Front. Plant Sci. 3:85. doi: 10.3389/fpls.2012.00085

FIGURE 11.6 Cell wall apposition (growth in thickness and area) seals off invading fungal threads.

Delayed active response (days)

Delayed active responses limit the spread of the pathogen. One important strategy is to repair wounds in the bark, through cork cell production and gum secretion. Lysozyme-like chemicals are also released and have an antimicrobial action. Salicylic acid may act as a signalling agent of subsequent infections and play a role in the plant's 'memory' of a particular pathogen. This is known as **systemic acquired resistance** and limits the severity of subsequent infections with that pathogen.

The responses of plants to pathogens are summarised in Figure 11.7.

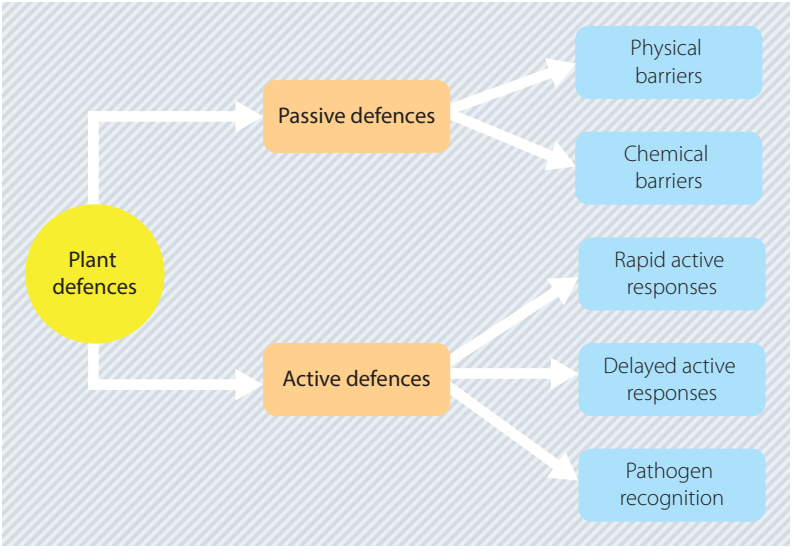

FIGURE 11.7 The types of responses by plants against pathogens

INVESTIGATION 11.1

Primary- and secondary source-investigation: Plant responses to pathogens

Critical and creative thinking

Ethical understanding

Personal and social capability

Fungi and viruses are common pathogens of plants in Australia. You are to investigate a specific pathogen of an *Australian plant* and the defence responses of that plant.

The outward signs of infection of plants by pathogens were discussed in Chapter 10. Changes in colour, abnormal growth, mottling of leaves and death of tissues can be explained in terms of direct or indirect damage inflicted by the pathogen and/or strategies used by the plant to destroy the pathogen or limit its spread. Examples of fungal and viral infections are shown in Figures 11.1 and 11.8 to 11.10.

FIGURE 11.8 Fungal invasion of a leaf

FIGURE 11.9 Viral infections often produce mosaic patterns on leaves.

AIMS

1. To investigate the response of a named Australian (native) plant to a named viral or fungal pathogen through practical investigation and secondary sources
2. To design and evaluate hypotheses related to responses of a plant to a pathogen
3. To assess the risks and consider ethical issues related to the selection and disposal of materials
4. To evaluate and/or modify your investigation in light of new evidence

FIGURE 11.10 Rose mosaic virus showing yellow patterns on leaves.

HYPOTHESIS

This is optional. You may like to make an informed prediction of what types of responses you will observe in a named disease, based on the information in Chapter 10. What types of changes will you expect to see in fungal and viral infections? Or you may wish to formulate your own research question based on this topic. (See page 3 for some guidelines.)

MATERIALS

List any materials that you may need to assist you with your research (e.g. magnifying glass, light microscope, Petri dishes).

RISK ASSESSMENT

Construct a risk assessment table, with the following considerations in mind.

1. The use of gloves is recommended, to prevent biological contamination of your skin and allergic reactions from contact with plant material.
2. The use of a face mask is strongly advised when examining plants infected with fungal material, to prevent inhalation of fungal spores.

»

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3 Protective eyewear must be worn.
4 A lab coat or apron will reduce the risk of getting fungal spores on your clothing.
5 All surfaces must be wiped down with alcohol after the experiment.
6 Your teacher will advise you on the safest means of disposal of infected plant material, to prevent transmission to non-infected plants in the environment.
7 All laboratory equipment used in this experiment should be thoroughly disinfected to remove pathogens. This includes microscope slides.
Develop your risk assessment table using the template below.

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?

METHOD

1 Work in pairs. Choose a viral or fungal pathogen of an Australian native plant. There are many good online resources to help you make your decision. Try to select something of interest from your local area.
2 Collect data on both the plant and the invading pathogen. It is up to you how you divide the workload, but one of you may like to research the plant and its responses, and the other may like to research the pathogen.
3 You may collect actual samples of the plant/pathogen. You may also use printed or online material (secondary sources). Conducting a literature review is also an option (page 9). The information you will be required to gather includes the following:
   a scientific and common name of the selected Australian plant
   b scientific and common name of the fungal or viral pathogen
   c diagrams or photographs of a normal plant and an infected plant . Include all parts of the plant, not just those visible above ground. This may include photos of the whole plant, parts of the plant or even images taken using light and electron microscopy. If you take photos of a plant outside the classroom, consider using a 'selfie stick' for hard-to-reach parts of the plant. You may wish to include flow charts, diagrams or other representations showing more subtle chemical and immune changes in your chosen plant (e.g. biochemical signalling).
   d an explanation of the changes the plant undergoes. You must link the cause (pathogen) to the effect (plant response).
4 Apply the CRAAP test to your sources (page 10).

The weblinks may be a good starting point for your investigation.

Weblink
Royal Botanic Garden Sydney

Weblink
Australian Government website

Weblink
Australian Native Plants Society

RESULTS

Construct a table to record your results. Use the table below as a guide.

PLANT NAME	NAME OF FUNGAL OR VIRAL PATHOGEN	DIAGRAMS/PHOTOS OF AFFECTED PLANT

DISCUSSION

1 Swap your research with another group and peer review each other's research in terms of:
   a reliability
   b accuracy
   c validity
   d relevance.
2 Describe how the pathogen enters the plant.
3 In what types of ways does the plant respond to infection?
4 Describe the effect of the pathogen on the plant.

CONCLUSION

The conclusion should refer back to the hypothesis and aim.

KEY CONCEPTS

- Plants respond to pathogens by both chemical and physical means.
- Passive defences include physical barriers and chemical barriers.
- Active defences include rapid and delayed responses.
- Rapid responses help to seal off any wounds and destroy pathogens quickly.
- Delayed responses limit the spread of an invading pathogen and involve complex chemical signalling.

CHECK YOUR UNDERSTANDING 11.1

1. An insect begins to feed on a plant's stem and breaches the epidermis with its mouthparts. Outline the steps the plant might take to seal the breach and deal with potential pathogens introduced by the insect.
2. Explain why vertically hanging leaves might be an advantage to a plant in an environment that is rich in potential pathogens.
3. What chemical response to pathogens closes stomata? Evaluate the usefulness of this response.
4. Justify the responsible disposal of pathogen-infected plant material used during school experiments.
5. A plant has discolouration on its leaves. What signs would you look for, or techniques could you use, to determine whether the pathogen is a fungus or a virus?
6. Imagine that you are the manager of a small plant nursery. A supplier has brought in a new batch of roses and placed them on display. You notice a day later that they are showing signs of a bacterial disease.
   a. What steps could you have taken to prevent these infected plants being exposed to other healthy plants?
   b. What procedures would you put in place to reduce the spread of plant pathogens in your nursery? Think of layout of the nursery, materials used in construction, and personnel routines.

11.2 Animal responses to pathogens

Every good general of the armed forces knows that a battle must be meticulously planned and organised. A leader who can anticipate possible moves by the enemy is able to prepare in advance and train the troops for scenarios they have not yet encountered. All soldiers have a rank and a specific role. If plan A fails, there is plan B and, if necessary, plan C.

With living organisms, the vast array and complexity of micro-organisms to which they are exposed presents a challenge to their defence mechanisms. Complex multicellular organisms such as animals (including humans) and plants have a multilayered system of defences to prevent infection and colonisation by potential pathogens. The immune system of animals is similar to an army: it is highly organised into different levels or ranks, and it is able to anticipate invasion by pathogens it has not yet encountered. There are multiple layers of defence, including physical and chemical responses, for when pathogens land on and/or breach the outer boundaries of the organism.

Lines of defence

The body's immune system consists of different levels of defence, as shown in Figure 11.11.

Innate immunity is present at birth and is genetically determined. Its responses to pathogens are non-specific, and include both physical and chemical barriers (first line of defence) as well as cellular responses (second line of defence).

1. The body's first line of defence against pathogens consists of barriers to entry. These barriers may be physical (such as skin), chemical (such as tears) or biological (such as sphincters).

2 When a barrier is breached, the second line of defence is activated. It involves a non-specific chemical and cellular attack on the pathogen, characterised by the process of **inflammation**.

Both these stages are non-specific – they act against any substance that is perceived as 'non-self' and a threat, including invading living organisms and organic and inorganic substances (such as a splinter in the skin, a transplanted organ or dust in the eyes).

Adaptive immunity is the third line of defence. This is a specific defence mechanism consisting of specialised cells that act if the pathogen persists in its invasion.

The third line of defence is discussed in Chapter 12.

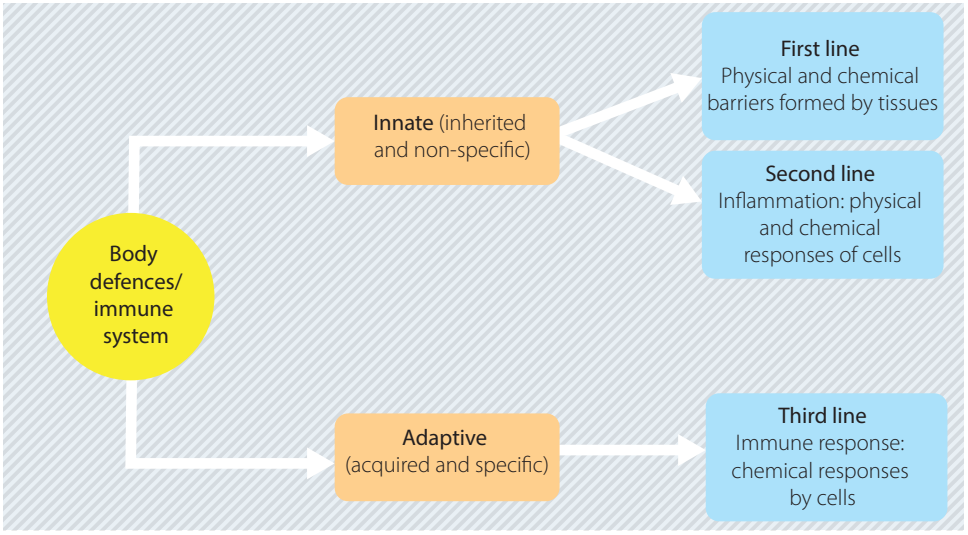

FIGURE 11.11 Organisational levels of the body's defences

The arrangement of the body has implications for the immune system

The outside parts of organisms are constantly exposed to the environment. The internal compartments that are closed off to the environment must remain **sterile** (free from microbial contamination). These include the thoracic, cranial and abdominal cavities, and the blood vessels of the cardiovascular system. The organism must prevent entry of pathogens to these spaces, or severe and overwhelming consequences may result. This is why a ruptured appendix or a penetrating wound to the chest is an emergency. It is also why organisms have developed elaborate mechanisms to prevent entry of pathogens to such spaces.

Think of the body as a plastic tube or pipe (Fig. 11.12). The inside tunnel (lumen) represents the digestive system, which is full of bacteria. Some of these bacteria are essential, such as our microflora,

senee sriyota/Shutterstock.com

FIGURE 11.12 A tube or pipe is a good scientific model for the body.

but some, such as *E. coli*, are harmful in large numbers. Both microflora and pathogens must be excluded from the body's sterile areas. The outside of the tube or pipe is exposed to environmental micro-organisms. The plastic itself represents the tissues of your body. The entire defence system of multicellular organisms is dedicated to prohibiting entry of micro-organisms to the 'plastic' itself. Tissues must be kept sterile. Keep this model in mind when you read further about the three lines of defence.

KEY CONCEPTS

- The body's defences are organised into three 'lines'.
- The first line of defence consists of physical and chemical barriers to pathogens.
- The second line of defence is initiated by chemical signalling from damaged tissue and consists of the inflammatory response and phagocytosis by white blood cells.
- The third line of defence is a specific defence by lymphocytes in response to chemical signals.
- Certain parts of the body must remain sterile and free of contamination by both pathogens and microflora.

CHECK YOUR UNDERSTANDING 11.2a

1. Explain why a surgeon sterilises the skin around where they plan to make an incision during an operation. Use your knowledge of the body's barriers and sterile areas.
2. In what ways are the body's defensive systems like an army?
3. A model of the body's defences is that of a hollow tube. In a group, brainstorm possible ways to modify the tube model of the lines of defence to make it a more accurate representation of the body's immune defences.
4. Apply your understanding of evolution by natural selection to explain how plants and animals have developed such complex and successful defences against pathogens.
5. Use the tube or pipe model to explain why a ruptured appendix is an emergency in terms of infection.

Components of the immune system

An **antigen** is any molecule that the body recognises as foreign and that triggers an immune response.

On the surface of cells in the body there are 'marker' molecules that identify the cell as belonging to the body ('self'). This protects the cells in the body from attack by its own immune system.

Pathogens that enter the body have a variety of chemical markers (antigens) on their surface; the immune system recognises these markers as not belonging to the body ('non-self'). In this way, the presence of antigens causes the immune response to activate, to destroy the pathogens.

It is not only pathogens that have antigens on their surface. Any foreign cell, cell fragment, protein debris or toxin produced by bacteria can also contain antigens. The venom of venomous snakes contains a number of antigens. In all these examples, the immune response is activated because the body recognises all these antigens as foreign (non-self) molecules.

When pathogens are successful in penetrating the barriers against entry into the organism, non-specific responses that are the 'second line of defence' are quickly activated to try to destroy the invaders before they can cause any damage to the body.

These second-line, non-specific defence adaptations include:

- inflammation
- phagocytosis
- fever (pyrexia)
- cell death to seal off the pathogens (granuloma formation).

These processes work together, ahead of and with the third line of defence if required, to defend the body against attack.

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INVESTIGATION 11.2

Secondary-source investigation: Christmas trees and asthma

Critical and creative thinking

Information and communication technology capability

Mould is a fungus. It is present everywhere in the environment. In homes, mould can grow in moist areas with poor ventilation. It is associated with health problems in humans and animals.

A strange phenomenon has been observed in the United States in an area near New Jersey. Since 1930, there has been an increase in the severity of symptoms of asthmatics during the Christmas season. In this investigation, you will examine the qualitative and quantitative data associated with this research.

AIMS

1. To conduct an investigation to gather valid and reliable secondary data on the incidence of respiratory disease at Christmas in Washington Township, New Jersey
2. To analyse and evaluate secondary data and information on the incidence of respiratory disease
3. To identify trends and relationships in the data and determine whether there is a link between Christmas trees and the increased incidence of respiratory disease
4. To determine what alternatives to live Christmas trees are available

Weblink
The Christmas tree allergy phenomenon

HYPOTHESIS

Write a suitable hypothesis for your investigation.

METHOD

Construct a method to test your hypothesis. Use the weblinks to start your research. Collect both quantitative and qualitative data.

Weblink
Christmas tree allergy: mould and pollen studies

RESULTS

Record your observations in a carefully formatted table.

DISCUSSION

1. Discuss your results, with particular attention to the link between live Christmas trees and the increased incidence of respiratory disease. Are non-living trees safer? Use data to support your answer.
2. Make recommendations based on your results.
3. Analyse the validity, reliability, accuracy and relevance of your sources.

Weblink
Mould in Christmas trees

CONCLUSION

Summarise your findings about the potential dangers of live Christmas trees, referring back to your aims and hypothesis.

The role of the lymphatic system

As blood circulates around the body, some of the plasma moves out of the capillaries into the tissues and becomes part of the tissue fluid. This tissue fluid then moves into a system of vessels known as the lymphatic system. The lymphatic system consists of lymph (a milky fluid), lymph nodes, lymph vessels, thymus, spleen, tonsils and adenoids (Fig. 11.13).

The lymph vessels form a one-way drainage system from all parts of the body back to a point near the heart, where the cleansed lymph fluid is drained back into the blood via the thoracic duct. The muscles that surround the vessels squeeze the fluid in one direction and valves prevent the fluid moving backwards.

At different points along the lymph vessels, structures called lymph nodes play an important part in the body's defence system.

During infections, pathogens and their products may enter the lymph fluid. The fluid is transported to the local lymph nodes, where substances including microbes, cellular debris and cancer cells are filtered out of the fluid before it continues its journey. One of the ways that infections may be detected is by the enlargement of the local lymph nodes. Changes in size, shape or texture of lymph nodes may help to pinpoint the site of an infection, as the drainage routes for the particular lymph node are known.

Therefore, swollen lymph nodes (glands) are a good indicator of the body's response to infection (Fig. 11.14). The lymph nodes also play an important role in the third line of defence, as you will learn in Chapter 12.

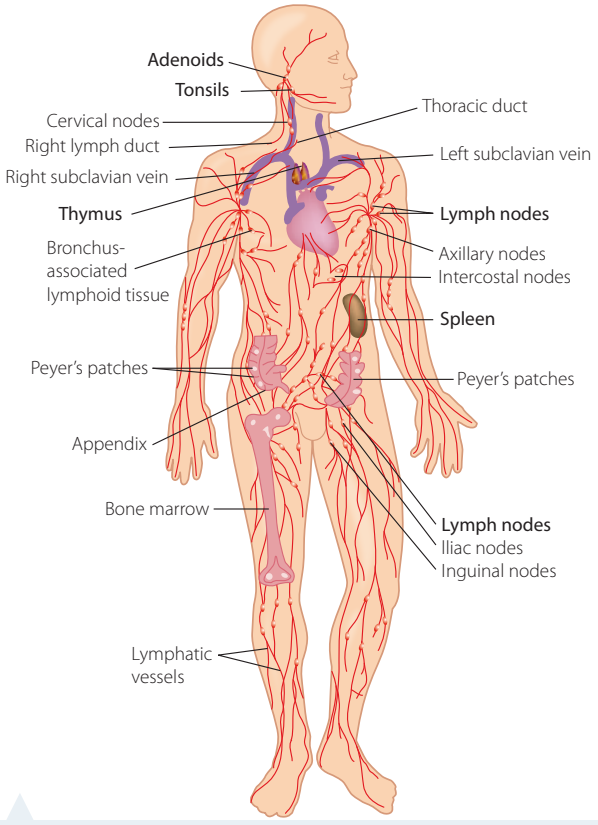

FIGURE 11.13 The lymphatic system consists of lymph vessels, lymph nodes and lymphoid organs such as the spleen and thymus.

FIGURE 11.14 Lymph nodes play an important role in defending the body. Swollen lymph nodes can be an indicator of infection.

White blood cells

White blood cells (leucocytes) play a pivotal role in the innate and adaptive responses to pathogens. There are different types of white blood cells, which differ in:

- size
- whether they contain granules in their cytoplasm (granular or agranular)
- the colour of their granules and cytoplasm (pink – eosinophilic, blue – basophilic)
- the shape of their nucleus (lobed or mononuclear).

Table 11.3 lists the functions and locations of the different types of white blood cells. Figure 11.15 shows their appearance. Note how they differ according to the criteria listed above.

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TABLE 11.3 Cells involved in chemical and physical responses to infection by pathogens (lymphocytes are not included)

CELL TYPE	CHARACTERISTICS	LOCATION
Mast cell	• Blood vessel dilation • Release of heparin and histamines • Recruitment of neutrophils and macrophages • Also involved in allergic reactions	Connective tissues and mucous membranes
Macrophage	• Phagocytosis of pathogens and cancer cells • Antigen-presenting cell	Migrates from blood vessels into tissues
Natural killer cell	• Kills tumour cells and virus-infected cells	Circulates in blood and migrates into tissues
Dendritic cell	• Antigen-presenting cell • Triggers adaptive immune response	Epithelial tissues, including skin, lung and tissues of the digestive tract. Migrates to lymph nodes upon activation
Monocyte	• Differentiates into phagocytic cells such as dendritic cells and macrophages	Stored in spleen. Moves to infected tissues through blood vessels
Neutrophil	• Most common white blood cell at site of trauma or infection • Releases toxins that kill or inhibit bacteria and fungi • Recruits other immune cells to the site of infection	Migrates from blood vessels into tissues
Basophil	• Defence against parasites • Releases histamines that cause inflammation • Responsible for some allergic reactions	Circulates in blood and migrates to tissues
Eosinophil	• Releases toxins that kill bacteria and parasites	Circulates in blood and migrates to tissues

Source: adapted from Lumen Learning Canada 2017, Boundless Biology, 'Innate immune response', https://courses.lumenlearning.com/boundless-biology/chapter/innate-immune-response/

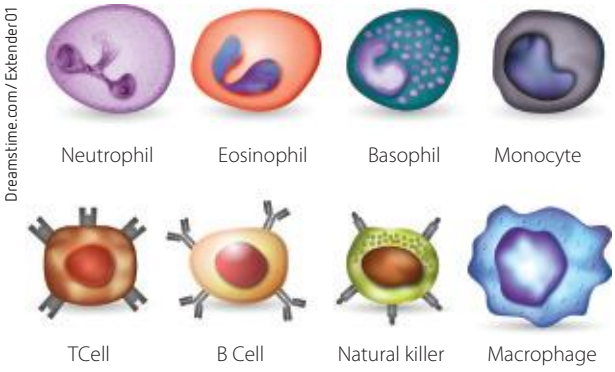

FIGURE 11.15 Cells involved in chemical and physical responses to infection by pathogens. The cells are not drawn to scale.

Figure 11.32 shows dendritic cells.

The microbiome: a natural barrier to infection

The human body is home to a large and varied population of microbes, living mainly on the skin, in the intestines, the colon, the mouth and the vagina in women. These microbes are known collectively as the **microbiome** of the body. The body supplies these microbes with the nutrients they require and the conditions they need to survive. In return, the presence of the microbiome inhibits the growth and multiplication of many pathogens that encounter the body, because the natural microbes out-compete the pathogenic ones. The microbiome thereby protects the body from many diseases.

If the conditions of the body change, and the balance of the microbiome is changed, the growth and multiplication of harmful pathogens may not be controlled, which can lead to an increase in their numbers and the development of disease. Of the many diseases that are caused when there is an imbalance in the microbiome of the body, one example is candidiasis (commonly known as thrush).

Candidiasis is caused by the fungus *Candida albicans*. This fungus (yeast) is part of the natural microbiome of the human body and is normally present on the mucous membranes of the female genital tract, mouth, respiratory tract and alimentary canal. The number of *C. albicans* is usually kept low by competition from other micro-organisms in the body's microbiome. If the natural balance of the microbiome is upset for any reason, the number of *C. albicans* increases and the disease candidiasis develops.

One of the ways in which the natural balance of the microbiome can be upset is by taking antibiotics to treat a bacterial infection. As well as reducing the number of pathogenic bacteria in the body, antibiotics can also reduce the number of bacteria in the natural population of microbiome. This allows *C. albicans* to multiply in an uncontrolled way and the disease candidiasis to become established.

Other factors that allow an increase in the number of *C. albicans* include:

- suppression of the immune system from inherited conditions, cancer treatment, and viruses such as HIV/AIDS
- diabetes mellitus – high sugar levels predispose to *Candida* overgrowth
- use of corticosteroids, which suppress immune surveillance
- hormones – higher oestrogen levels in the second and third trimesters of pregnancy can predispose to thrush
- use of oral contraceptives – associated with higher oestrogen levels
- general illness – can lead to suppression of the immune responses
- intravenous drug use (such as heroin).

Physical barriers against infection

Worksheet
First line of defence in the immune system

Every day our computers are vulnerable to cyberattack. One of the most common ways to protect them is to have an active **firewall** program that denies entry to common computer viruses, such as the 'ransomware' viruses that plague businesses and institutions around the world. Similarly, the most effective way to deal with the threat of a pathogen is to deny it entry to the body. Living things have an array of natural physical firewalls or **barriers** to keep pathogens out.

Physical barriers are structures that the body uses to restrict entry to pathogens, by making it difficult for the pathogen to adhere to cells or to penetrate tissues.

Table 11.4 lists some of the ways in which pathogens can enter the body.

TABLE 11.4 Routes of entry for pathogens into the body

ROUTE OF ENTRY	MODE OF TRANSMISSION	EXAMPLES OF PATHOGENS	EXAMPLES OF DISEASES
Mucosal surfaces			
Airway	Inhaled droplets	Influenza virus *Neisseria meningitides*	Influenza Meningococcal meningitis
Gastrointestinal tract	Contaminated water or food	*Salmonella typhi* Rotavirus	Typhoid fever Diarrhoea
Reproductive tract	Physical contact	*Treponema pallidum*	Syphilis
External epithelia			
External surface	Physical contact	*Tinea pedis*	Athlete's foot
Wounds and abrasions	Minor skin abrasions Puncture wounds Handling infected animals	*Bacillus anthracis* *Clostridium tetani* *Pasteurella tularensis*	Anthrax Tetanus Tularaemia
Insect bites	Mosquito bites (*Aedes aegypti*) Tick bites Mosquito bites (*Anopheles*)	Flavivirus *Borrelia burgdorferi* *Plasmodium* spp.	Yellow fever Lyme disease Malaria

Source: adapted from Janeway CA, Travers P, Walport et al 2001, *Immunobiology: The Immune System in Health and Disease*, 5th edn, New York: Garland Science

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Epithelial tissues line all the major internal cavities of the body and include the **skin** and **mucous membranes**. Mucous membranes are the moist pink tissues lining the entry points of the body, such as the lining of the mouth, the nasal cavity, the conjunctiva and the entrances to the digestive and genitourinary systems. The skin and the mucous membranes have a range of strategies to restrict pathogen entry.

Skin

The skin is classified as epithelial tissue. It consists of three layers: the outer **epidermis**, the underlying **dermis** and the lower **hypodermis** (or subcutaneous tissue) (Fig. 11.16). Skin is well supplied by blood, which contributes to its effectiveness as a barrier to disease by providing early access for white blood cells, red blood cells and platelets to any wound.

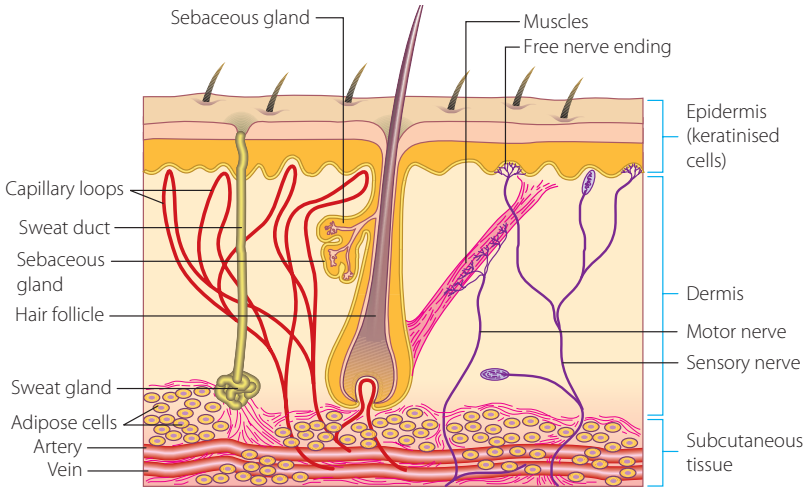
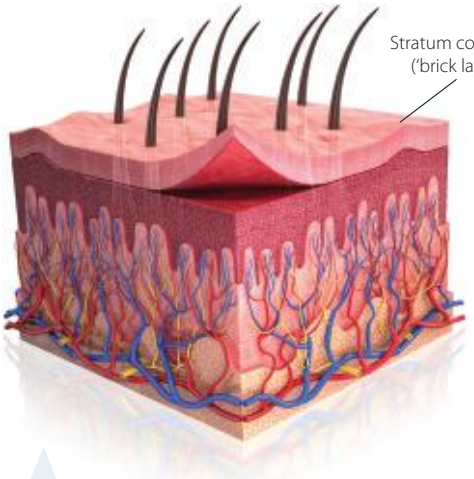


FIGURE 11.16 The skin forms a physical barrier against pathogens.

The epidermis consists of sheets of cells covered by a special barrier, called keratin. Keratin is a waterproof protein that provides an extra layer of security against pathogen entry. Keratin is secreted by skin cells known as keratinocytes. Keratin separates the organism from its environment very effectively, as it is mechanically tough and resistant to degradation by bacterial enzymes.

The upper layer of the epidermis, or *stratum corneum*, consists of a layer of flattened and dead skin cells in a type of 'bricks and mortar' arrangement (Fig. 11.17). This forms an effective physical barrier against pathogen entry. As dead cells exfoliate (die and flake off), they take pathogens with them.



Science Photo Library/Alamy Stock Photo

FIGURE 11.17 The 'bricks and mortar' structure of the upper layer of the epidermis makes skin an excellent physical barrier to pathogens.

When there is loss of integrity (completeness) of the skin barrier due to wounds or burns, the body has special processes to seal the site as quickly as possible:

1. inflammation – this will be discussed in more detail on page 385.
2. proliferation – new cells multiply rapidly to seal the wound
3. maturation – cells mature and complete the new barrier.

Mucous membranes

Many of the internal cavities of the body are continuous with the external environment. This means they are directly exposed to pathogens on a daily basis. These cavities are lined with a special type of epithelial tissue, called a mucous membrane, that forms a barrier to pathogen entry. The number of layers and shapes of cells depends on the location. Mucous membranes are typically pink, moist lining tissues, such as the lining of our mouth and nose (Fig. 11.18). They are found in the digestive (alimentary), respiratory and genitourinary systems.

The following features of mucous membranes restrict pathogen entry.

- **Cell junctions** between epithelial cells are designed to anchor them together more effectively, increasing cohesion and restricting access to pathogens.
- Many epithelial cells are lined with tiny hair-like structures called cilia, which beat in a coordinated way to remove particles from the respiratory system (Fig. 11.19) – this is known as the muco-ciliary escalator. It is this escalator that is damaged by cigarette smoke and contributes to the typical 'smoker's cough'.
- They are composed of sheets of cells that are constantly growing and moving upwards to replace surface cells lost due to wear and tear or pathogen attack.
- They secrete a number of protective substances such as mucus, lysozyme and immunoglobulins (antibodies).

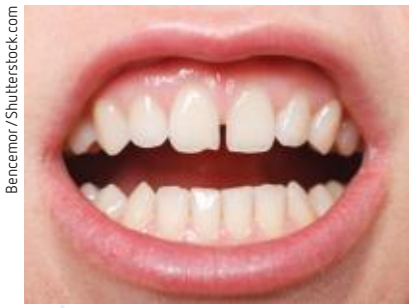


FIGURE 11.18 Mucous membranes line entry points to the body, such as the mouth.

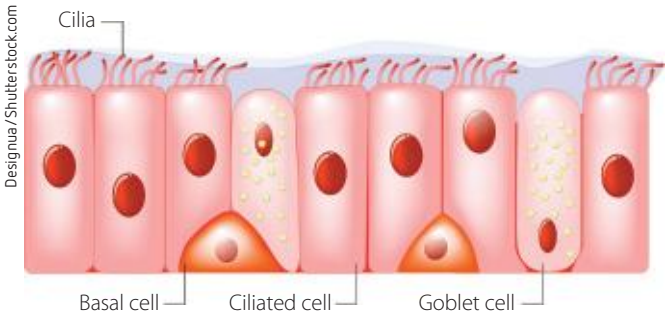


FIGURE 11.19 Cilia wave in a coordinated way to expel foreign material.

Tight junctions

Blood vessels are lined internally by **endothelial** cells. These cells have special ways to adhere tightly to each other, which helps prevent the entry of pathogens from infected tissue into the blood vessels (Fig. 11.20). If pathogens enter the bloodstream, they can travel to distant sites and set up new centres of infection. The presence of bacteria in the bloodstream is known as **bacteraemia**.

The **blood–brain barrier** is a particularly important example of the effectiveness of tight junctions. It is formed by brain endothelial cells connected by tight junctions, and restricts the diffusion of microscopic objects such as bacteria into the brain.

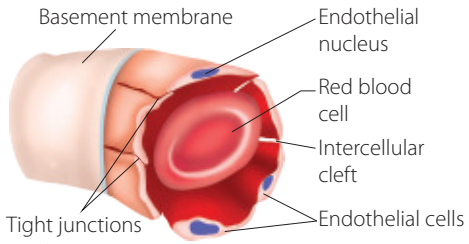


FIGURE 11.20 Tight junctions between endothelial cells form a physical barrier between the extracellular tissues and the bloodstream.

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Mucus

Mucus is a slippery substance secreted by cells that line the mucous membranes. Mucus protects the linings of the body by trapping foreign substances such as pathogens, dust and pollen. Increased mucus production in the respiratory tract (Fig. 11.21) is often a sign of ill health, as the body attempts to flush away an invading pathogen. A runny nose and cough may result. Green mucus can indicate the activity of white blood cells, as they secrete iron-containing enzymes to deal with pathogens.

Mucus also prevents the entry of pathogens through the cells lining the alimentary canal. Cervical mucus plugs can guard the uterus against entry by pathogens during pregnancy. Some intestinal mucus has been shown to contain substances that inhibit the replication of viruses, such as rotaviruses.

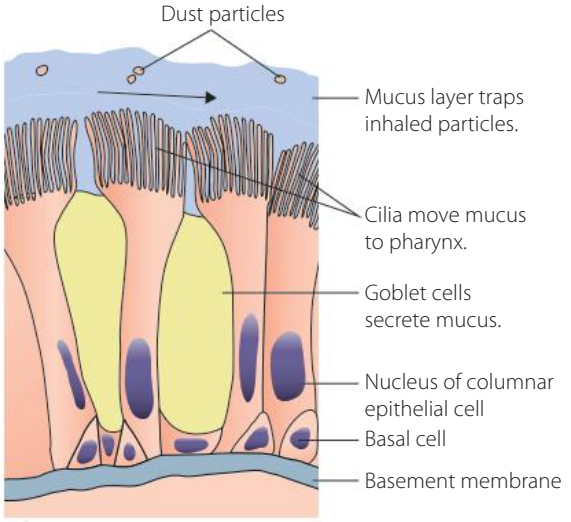


FIGURE 11.21 Mucus is produced by goblet cells that form part of the epithelial layer lining the respiratory tract.

Peristalsis

The alimentary canal is a long tube through which food moves from the mouth to the anus. The wall of this tube consists of several layers, one of which is smooth muscle. Smooth muscle is not under voluntary control but acts automatically. This muscle contracts in a coordinated way, known as peristalsis, to move food in one direction only. Stasis (lack of movement) of the intestines can lead to intestinal bacterial overgrowth. Bacterial overgrowth occurs because the bacteria have an opportunity to proliferate (reproduce).

Sphincters

A sphincter is a circular muscle that maintains constriction of a natural body passage or orifice and relaxes as required by normal physiological functioning. Sphincters are found in many parts of the body (Fig. 11.22), including:

- the lower oesophageal sphincter – between the oesophagus and the stomach
- the pyloric sphincter – between the stomach and the duodenum
- the ileocaecal sphincter – between the ileum (final part of the small intestine) and the large intestine
- the urethral sphincters – prevent the release of urine from the bladder unless done so voluntarily
- the sphincter of Oddi – ensures one-way flow of digestive juices from the common bile duct (from the gallbladder and pancreas) to the duodenum.

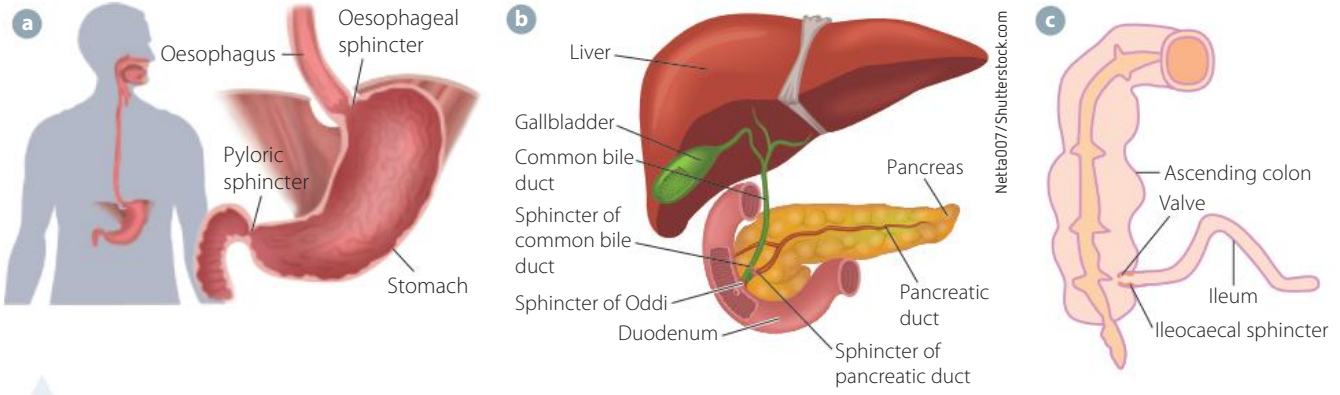


FIGURE 11.22 Sphincters help to physically seal off compartments in the body, to reduce the likelihood of pathogen invasion: **a** oesophageal and pyloric sphincters; **b** sphincter of Oddi and sphincter of pancreatic duct; **c** ileocaecal sphincter.

KEY CONCEPTS

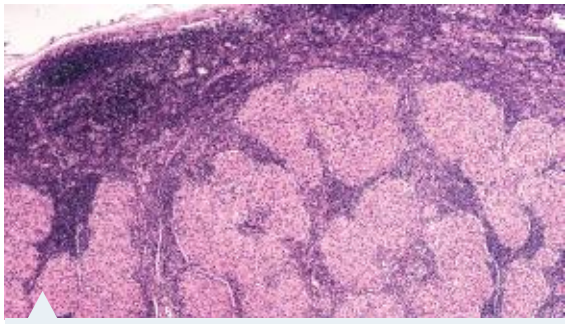
- Antigens trigger an immune response.
- Antigens may come from pathogens or from non-living material.
- Innate immune responses include inflammation, phagocytosis, fever and cell death to seal off pathogens.
- The lymphatic system consists of many tissues and organs involved in immune responses to pathogens.
- White blood cells play different roles in the response to pathogens.
- The human body hosts a large number of micro-organisms known as the microbiome, which assist in the maintenance of good health.
- Physical barriers restrict entry by pathogens, by making it difficult for them to adhere to or penetrate tissues.

CHECK YOUR UNDERSTANDING 11.2b

1. Why is it important that the body is able to detect and respond to the presence of antigens?
2. Outline the advantages of having a lymphatic system.
3. When viewing blood samples under a light microscope, what kinds of features allow scientists to distinguish between the different types of white blood cells?
4. What is the microbiome? How does the microbiome provide a barrier to pathogens in the human body? Outline an example of an infectious disease caused by an imbalance in the microbiome.
5. When a person takes an oral antibiotic for a number of weeks, they sometimes develop diarrhoea. It is often recommended by doctors that they eat yoghurt or other probiotics during their antibiotic course. Justify the use of probiotics in this situation.
6. How do the following prevent the entry of pathogens?
   - **a** skin
   - **b** peristalsis
   - **c** mucus
   - **d** sphincters
   - **e** cilia
7. Justify the use of a Band-Aid® when you have a wound on your skin from a scrape or a cut. What is it temporarily taking the place of?

Physical responses to infection

Sometimes cells die to seal off an area of tissue that is infected and is not being successfully defended by the body. If the infected cells are surrounded by a wall of dead cells, this prevents the infection from spreading to other areas and infecting them. This wall of dead cells forms a capsule known as a **granuloma** (Fig. 11.23). The cells inside the granuloma then die, causing the destruction of the pathogens that are infecting them. The debris inside the granuloma is destroyed by macrophages that have surrounded the 'walled-off' area. The bacteria that cause tuberculosis and leprosy (*Mycobacteriam* spp.) typically cause granuloma formations. Granulomas in the lungs were referred to as 'tubercles' (thus the name 'tuberculosis'). Granuloma formation is part of the second line of defence.



David A Litman/Shutterstock.com

FIGURE 11.23 Micrograph of a granuloma

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Vomiting and diarrhoea

Vomiting (**emesis**) is a reflex action coordinated by the vomiting centre (chemoreceptor trigger zone) of the brain. It happens in response to many different signals, one of which is the presence of pathogens in the gut (gastroenteritis). It is the body's way of expelling harmful substances. Interestingly, hypersalivation occurs before vomiting in order to protect your tooth enamel from stomach acid. It is thought that dogs and cats may chew on grass when feeling ill because grass acts as a natural emetic to help them vomit and expel harmful substances.

At the other end of the digestive system, **diarrhoea** expels micro-organisms quickly from the gastrointestinal system.

Increased urination

When the bladder lining is attacked by a pathogen, a common response by the body is inflammation (*cystitis*) and the need to pass frequent small amounts of urine (*pollakiuria*). This is thought to be a response by the body to help flush out pathogens (Fig. 11.24).

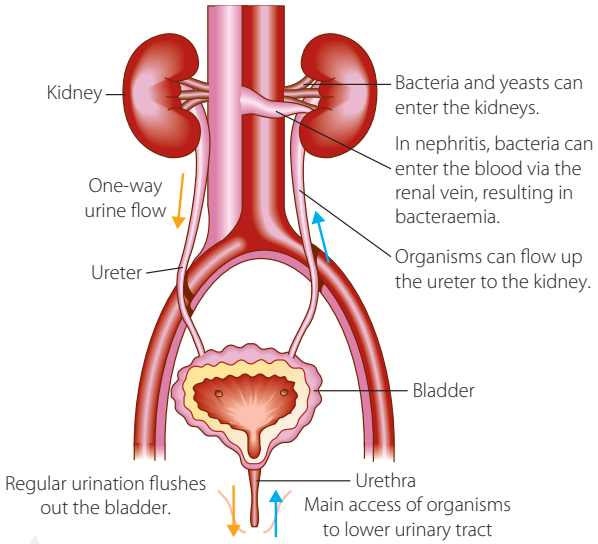


FIGURE 11.24 The one-way flushing action of urine assists in removing bacteria.

INVESTIGATION 11.3

Secondary-source investigation of urinary tract infections

Critical and creative thinking

Information and communication technology capability

Urinary tract infections (UTIs) are common in humans, particularly in the very young and in older men and women, as well as in other animals such as dogs and cats. They are commonly caused by bacterial pathogens. A person may be at greater risk of developing an infection of the urinary tract if there are other disease processes at work in their body. In this investigation, you will research the predisposing factors, symptoms and response of the body to the infection as well as the management and treatment of this type of infection.

AIMS

1. To pose questions regarding the development of UTIs
2. To make predictions about the risk factors involved in UTIs
3. To analyse and evaluate evidence from secondary sources
4. To communicate scientific and technological understanding of UTIs

HYPOTHESIS

Write a suitable hypothesis for your investigation. It may relate to the types of people who are at greater risk of UTIs or specifically to the factors that increase the risk of UTIs.

METHOD

1. Use printed and online secondary sources to gather secondary qualitative and quantitative data on the incidence and/or prevalence of UTIs, in either humans or another species (for example, dogs, cats).
2. Find a suitable species-specific diagram of the urinary tract so that you are familiar with the following parts: kidneys, ureters, bladder, urethra.
3. Identify the common pathogens that are most likely to cause UTIs. Where do these pathogens come from?

»

4 By what mechanism do these pathogens gain access to the upper urinary tract?

5 Describe the physical and chemical barriers that prevent bacteria colonising the urinary tract.

6 How does the urinary tract respond to the presence of a pathogen?

7 What signs and symptoms are produced as a result of the body's response to the presence of the pathogen?

8 Why are the very young and the very old more predisposed to this type of infection? What aspects of their body or behaviour allow it to happen more often? If you have chosen to research UTIs in cats or dogs, what types of factors increase the risk of this type of infection in these animals?

9 How are UTIs managed? Describe how these strategies aim to restore the usual chemical and physical defences against pathogens.

10 Use the weblink as a starting point for your research. Construct a list of other sources and references in the appropriate manner.

Weblink
Urinary tract infections

RESULTS

Record your findings in a carefully formatted table.

DISCUSSION

Make a general recommendation regarding ways to reduce the incidence of UTIs. Use your data to support your recommendation.

CONCLUSION

In what way does management of UTIs aim to restore the normal physical and chemical defences of the urinary tract against pathogens?

Wound healing

When there is a breach in the body's barriers, the tissues are exposed to environmental pathogens and the microbiome of the skin. There may be bleeding, if blood vessels have been damaged in the process. The priorities of wound healing are to:

- stop the bleeding (haemostasis) to maintain normal blood pressure
- confront pathogens, to prevent infection
- heal and repair the wound, to re-establish the barrier.

These stages overlap. Stopping the bleeding is the first priority for the body. To do this, the blood vessels contract (vasoconstriction) and a **platelet** plug is formed. A protein called **fibrin** forms a mesh, to trap more platelets and form a **clot** to seal the wound. The inflammatory response then begins.

KEY CONCEPTS

- Physical defences against infection include barriers and biological functions such as peristalsis.
- Barriers such as the skin, epithelium, mucus, tight junctions, peristalsis and sphincters all play a role in limiting access of pathogens to the body.
- Peristalsis causes the one-way movement of food matter from the mouth to the anus and prevents faecal matter moving backwards into more sterile compartments of the gut.
- Physical responses to infection include the following.
  - Vomiting and diarrhoea remove potential pathogens and their toxins quickly from the gut.
  - The one-way flow of urine flushes pathogens from the urinary tract and reduces the likelihood of pathogens moving into sterile areas such as the bladder and kidneys.
  - Wound healing reseals the physical barriers against infection by pathogens.

CHECK YOUR UNDERSTANDING 11.2c

1. A patient presents to a hospital with vomiting and diarrhoea after eating spoiled food. Explain why the body initiates this physical response to the presence of a pathogen in the gut.
2. Wound healing is a process that takes a lot of resources and energy to complete, and yet it is done relatively quickly in the absence of infection. Justify the 'cost' to the body in terms of the benefits of fast healing of breaches to the skin and mucous membranes.
3. Having to blow your nose a lot when you have a cold is often an inconvenience. Why do the membranes lining your nose respond to pathogens by hypersecretion of mucus from goblet cells in your nasal epithelium?
4. Assess the advantages and disadvantages of granuloma formation during a bacterial infection.
5. Cigarette smoke can cause loss of cilia on the epithelial cells lining the respiratory tract. Explain why smokers are more predisposed to bacterial infections of the respiratory system than non-smokers.
6. Bacterial infections of the gut are often accompanied by reduction or loss of peristalsis (intestinal motility disorder). Assess the importance of re-establishing normal gut movements as soon as possible in order to manage the infection.

Chemical defences against infection

In combination with physical barriers, a number of non-specific chemicals are secreted by epithelial tissue to further prevent pathogens entering the internal environment of the body. These secreted chemicals are found in mucus, saliva, tears, urine, stomach acid and natural antimicrobial substances, as well as body fluids.

Urine

Urine is sterile until it reaches the lower **urethra**. The precise mechanisms by which the urinary tract maintains sterility are not well understood. New research suggests that a microbiome may exist inside the bladder. When an animal or human is not urinating, there is a window of opportunity for pathogens to ascend the lower urinary tract (urethra) from the lower urethra, penis and vagina. Females have a shorter urethra than males and therefore are at greater risk of lower urinary tract infections. Faecal bacteria such as *E. coli* are a particular threat, as are normally **commensal** organisms such as *Staphyloccoccus* spp. found on the skin in this area. In bacterial **cystitis** (bladder infection), organisms make it all the way up the urethra into the bladder. The flushing activity that takes place during **micturition** (urination) assists in keeping pathogens away from the bladder.

The following chemical components of urine help defend against pathogens:

- antimicrobial peptides (AMPs) secreted by the cells lining the urinary tract to prevent binding of bacteria to epithelial cells and lyse (or break down) bacterial cells
- pH of normal human urine within the range of 4.5 to 8, with an average of 5–6 (slightly acidic). **Phagocytes** such as neutrophils work best when urine is alkaline. Doctors may prescribe urinary alkalinisers for urinary tract infections.

Sebum and sweat

Sebum is an oily material secreted by sebaceous glands. Its purpose is to waterproof and lubricate the skin. The pH of skin is normally around 5.5 (acidic), because of the presence of lactic acid, amino acids and fatty acids in sweat and sebum. Lysozyme is secreted in perspiration and lyses or breaks down bacterial cell walls (Fig. 11.16).

Saliva

Saliva is produced by the salivary glands. It is a complex mixture of water, mucus, electrolytes, enzymes such as **amylase**, and antimicrobial substances such as lysozyme and **immunoglobulin A (IgA)**. Saliva has a flushing action against microbes as well as chemical activity against them due to antimicrobial molecules contained within it, such as IgA and other antimicrobial peptides (AMPs).

Tears

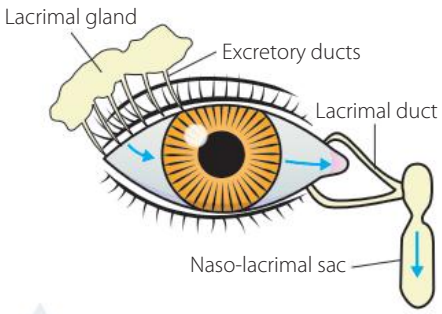


FIGURE 11.25 Anatomy of lacrimation: tears are produced by the lacrimal gland, and drain via the lacrimal duct into the nasal cavity.

Lacrimal glands (Fig. 11.25) produce tears. The glands along the eyelid edges also secrete a sebum-like substance that contributes to the tears and has antimicrobial properties. The production of tears is called **lacrimation**. This process produces a tear film that covers the **cornea** and **conjunctiva**. Goblet cells in the conjunctival epithelium secrete mucus, which helps distribute the film evenly.

The tear film contains the following chemical substances, which have antimicrobial properties:

- lysozyme, lactoferrin, lipocalin
- AMPs
- complement
- IgA
- mucins.

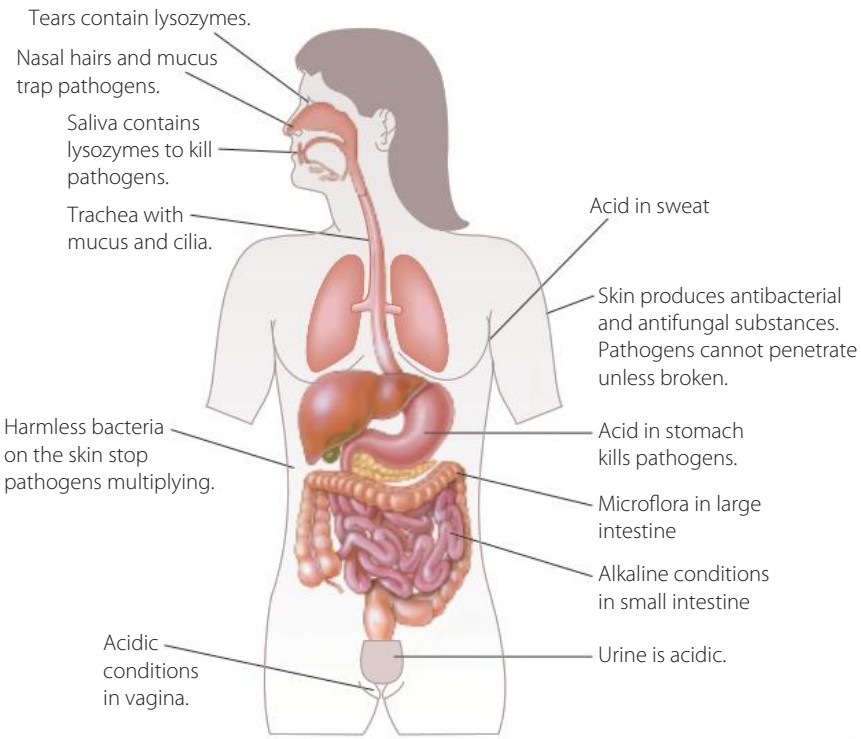
Gastric (stomach) secretions

The parietal cells lining the stomach (gastric) wall secrete hydrochloric acid. This creates highly acidic environment (pH 1–2) in the stomach, which discourages the growth and survival of microbes. In addition, the enzyme pepsin, secreted by the chief cells of the stomach wall, is thought to play an antimicrobial role. However, many bacterial pathogens, such as *E. coli*, *Salmonella typhimurium* and *Helicobacter pylori*, can circumvent the acidic conditions of the stomach by developing adaptive mechanisms that allow these bacteria to survive in acidic environments. Also, the act of eating may raise the stomach's pH above the threshold needed to destroy these bacteria. Therefore, these bacteria may survive acidic stomach conditions and pass into the intestinal tract, where they can cause **gastroenteritis**.

As food moves from the stomach to the duodenum, there is a rapid change in pH to around 6. This is due to the highly basic nature of bile released into the lumen of the duodenum. This is another limiting factor to pathogen growth.

A summary of chemical barriers is shown in Figure 11.26.

FIGURE 11.26 The first line of defence: barriers to the entry of pathogens



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KEY CONCEPTS

- Chemical defences against infection form part of the body's strategies to keep out pathogens.
- The antimicrobial substances in urine, as well as its pH, help create conditions that are unfavourable for the growth of pathogens.
- Sebum, sweat, saliva and tears all contain antimicrobial substances.
- Gastric acid produces a low pH in the stomach, which inhibits the growth of most pathogens.

CHECK YOUR UNDERSTANDING

11.2d

1. Describe the mechanisms by which urine uses chemical defences against pathogens.
2. Assess the claim that crying is a chemical response to the possible presence of a pathogen.
3. How does the stomach discourage the growth and survival of microbes?
4. If the fatty acids in sebum are bacteriostatic, why do pimples occur? Research secondary sources to gather and analyse the reasons for pimple formation.
5. Would people who regularly take antacids for reflux be at a greater risk of stomach infections due to the alteration in stomach pH?
6. Animals commonly lick their wounds after an injury. Justify the use of this response strategy in terms of chemical defence against the presence of pathogens.

Inflammation: a chemical response

Inflammation is a chemical response that helps wound repair and leads to pathogen destruction. The five cardinal signs of the inflammatory response are (by their latin names):

- *dolor* – pain due to the release of chemical mediators of inflammation
- *calor* – heat due to the increase in microcirculation
- *rubor* – redness associated with increased microcirculation
- *tumor* – swelling (oedema) as fluids move from the **intravascular space** to the **extracellular space**
- *functio laesa* – loss of function due to pain and swelling.

Worksheet
Second line of defence

The inflammation response is a non-specific defence mechanism and occurs at the site of infection. When the cells are challenged by pathogens or damaged in some way, they release chemical 'alarm signals'. These can be **histamines** (trigger vasodilation and increase vascular permeability), **bradykinin**, **serotonin** and **prostaglandins** (associated with the pain and fever of inflammation). These chemicals cause the capillaries to dilate, increasing the blood flow to the site of infection or injury and causing the area to become red, hot and swollen, painful and sometimes less mobile.

These chemicals also increase the permeability of the blood vessels, which allows certain white blood cells to move from the blood into the tissues to attack the invading pathogens (Fig. 11.27). Phagocytes are a special type of white blood cell. They include **neutrophils**, **macrophages** and **dendritic cells**. They are attracted to the site of inflammation by the presence of chemicals known as **chemotactic factors**.

Phagocytes are described in more detail in Chapter 12.

Chemicals that increase the body's temperature are released (**endogenous pyrogens**). This inhibits the growth of pathogens, inactivates some enzymes and toxins made by the pathogens, and increases the rate at which the biochemical reactions occur in the body.

When the pathogens are destroyed, they are removed along with any toxins, and the tissues are repaired.

The presence of a pathogen is not necessary for the body to respond to invasion. When damage occurs to tissues but the wound is sterile, **damage-associated molecular patterns (DAMPS)** are released by damaged cells. This sends a chemical signal to the surrounding tissue to initiate an inflammatory response. Afterwards, cellular replacement to restore damaged tissue occurs.

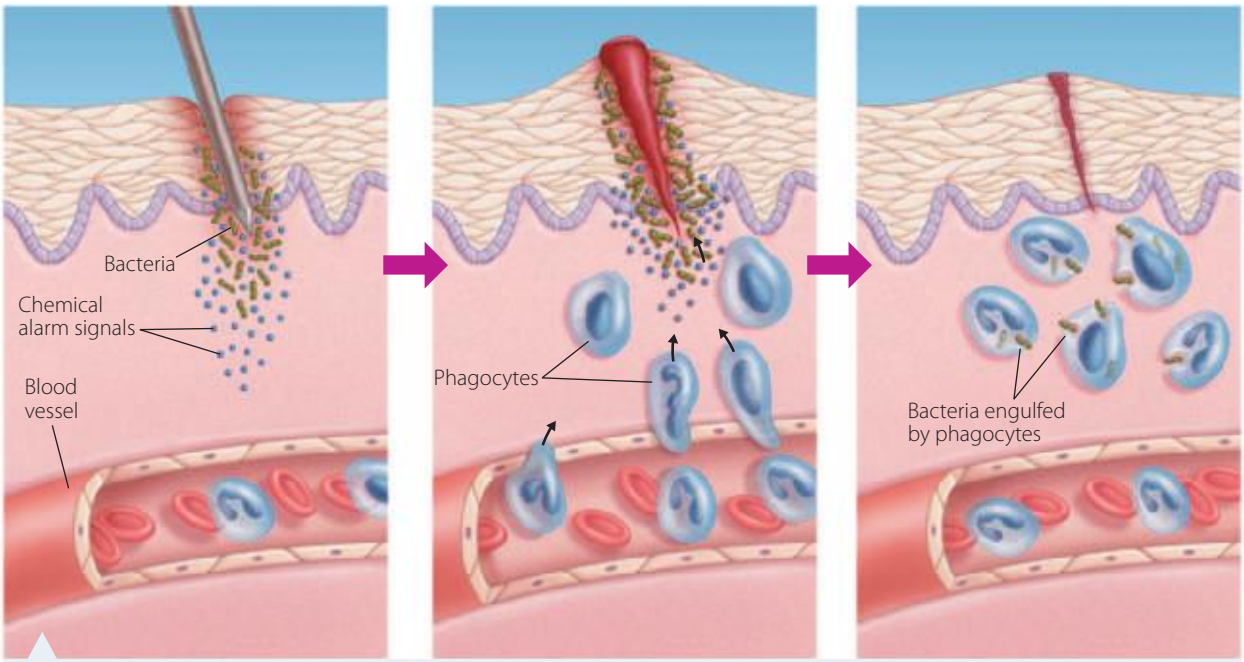

FIGURE 11.27 The inflammatory response: chemical signals at the site of bacterial entry attract white blood cells.

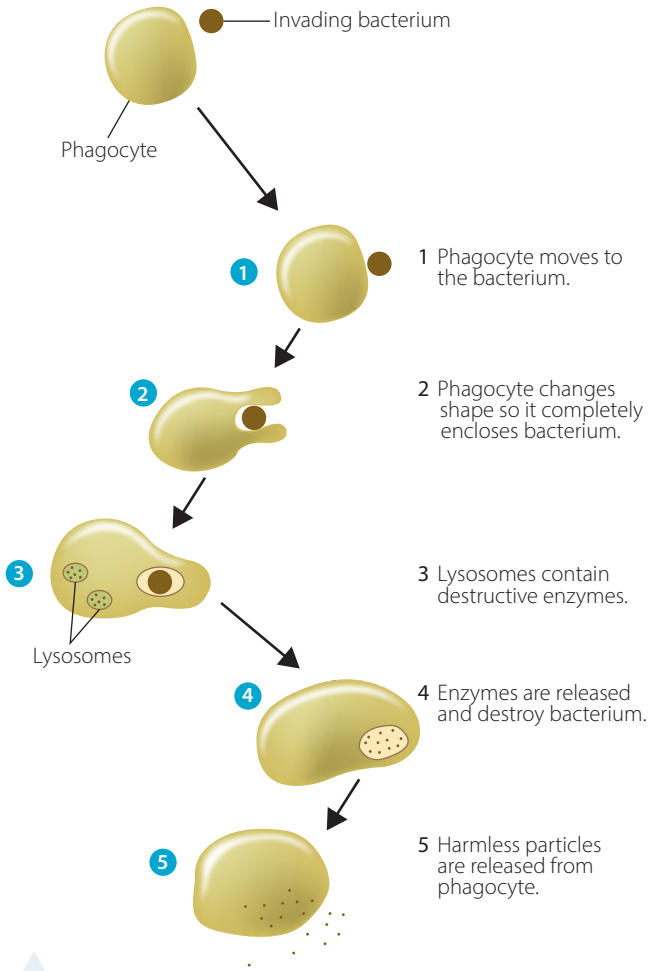

FIGURE 11.28 The process of phagocytosis

Phagocytosis: a response to chemical signalling

Phagocytosis is the process by which phagocytes change their shape so they can surround a foreign particle, such as a bacterium, and completely enclose it (Fig. 11.28). Once the foreign particle is inside the cell, enzymes are released to destroy it.

Phagocytes are specialised white blood cells or leucocytes. The main types of phagocytes are:

- neutrophils
- monocytes/macrophages
- dendritic cells
- natural killer cells.

Phagocytosis is not always successful, as pathogens can sometimes repel the phagocytes and may escape before being completely destroyed. In severe, overwhelming infections, immature neutrophils show toxic changes (and are referred to as 'toxic neutrophils') when seen in a blood smear. This is usually due to bacterial toxins circulating in the blood (and is a sign of septicaemia).

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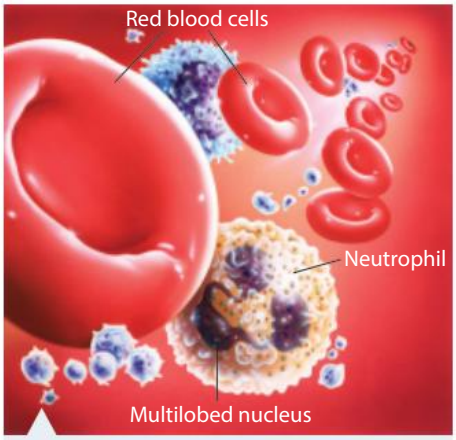
Neutrophils

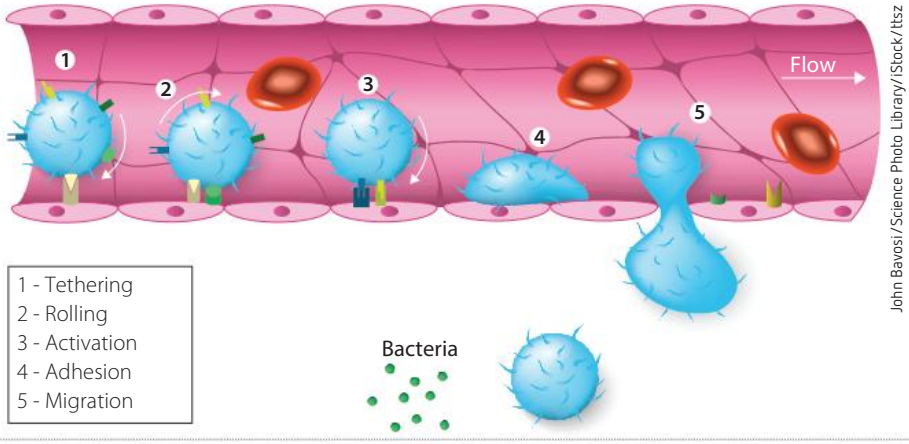
All phagocytic cells, including neutrophils (Fig. 11.29), originate in the bone marrow. They are capable of deforming and squeezing between the endothelial cells lining the local capillaries, to move from the blood to the tissues (a process known as diapedesis; Fig. 11.30). Neutrophils are the first to move to the site of the infection to inactivate pathogens. Neutrophils are short acting and then self-destruct after a few days. They are used by the body to fight acute (short, severe) infections. An increase in circulating neutrophils in the blood (neutrophilia) is indicative of an active site of inflammation somewhere in the body.



Science Photo Library/John Bavosi

FIGURE 11.29 A neutrophil is a large white blood cell with a multilobed nucleus.

It was once thought that neutrophils used substances such as peroxides in their vacuoles to destroy bacteria and fungi. It is now known that a complicated series of electron and ion flows occur, with movement of ions into the vacuole of the phagocyte, creating conditions that result in the death of microbes, which are then digested by enzymes such as **proteases**.



John Bavosi/Science Photo Library/iStock/ttsz

FIGURE 11.30 Diapedesis: migration of a neutrophil through a capillary wall

Monocytes

Monocytes circulate in the blood until attracted to inflamed tissue. They migrate through capillary walls to the inflamed tissue, where they undergo a transformation into macrophages and dendritic cells (Fig. 11.31).

The name 'macrophage' derives from the Greek terms for 'big eater'. Long-lasting phagocytes can either stay in the tissues or travel from the blood vessels into the infected tissues. They are used by the body to fight chronic (long-lasting) infections. After the macrophage has destroyed the foreign particle, parts of the antigen are displayed on the surface of the macrophage (antigen presentation).

When damage occurs to tissues, monocytes are recruited to the tissue from the blood. On the surface of monocytes are toll-like receptors (TLRs), which recognise specific pathogen-associated molecular patterns (PAMPs) released from bacterial cells. Monocytes quickly differentiate into macrophages and dendritic cells. They remove microbes, lipids and dying cells through the process of phagocytosis.

Dendritic cells also act as antigen-presenting cells. These dendritic cells, together with macrophages, act as 'bridges' between the innate and adaptive immune systems. You will see why this is important when you study the third line of defence. Interestingly, monocytes may also differentiate into osteoclasts ('bone breakers'), which are cells responsible for the maintenance, repair and remodelling of bone.

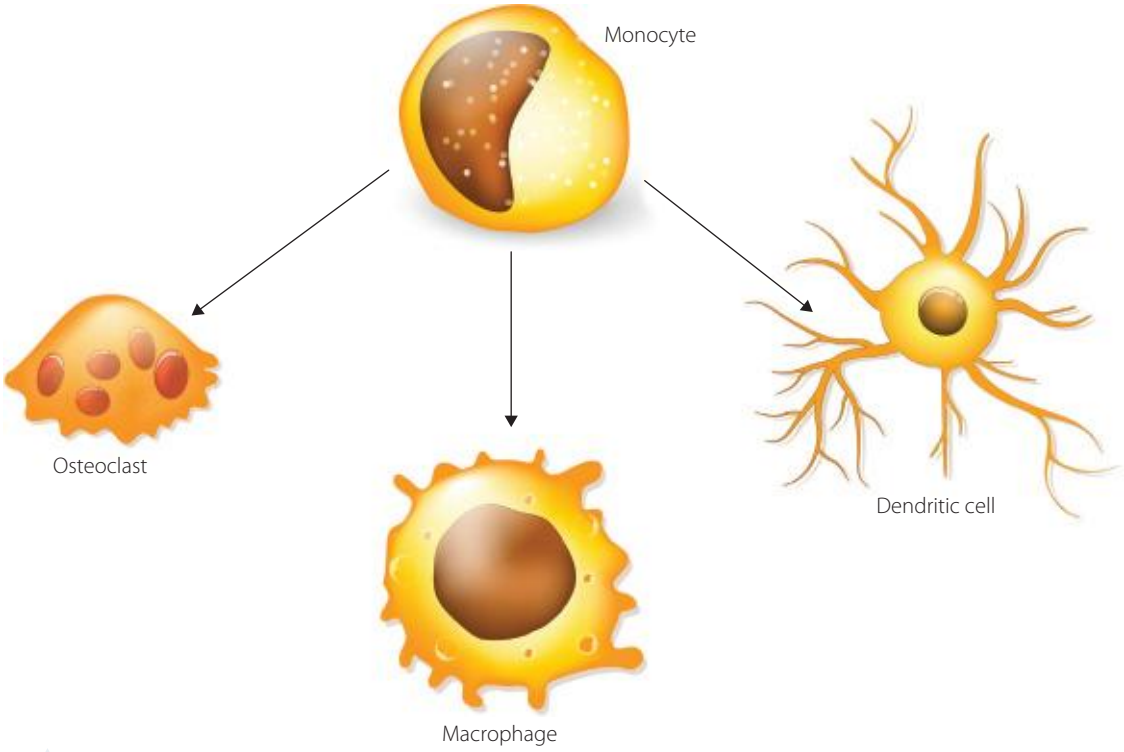
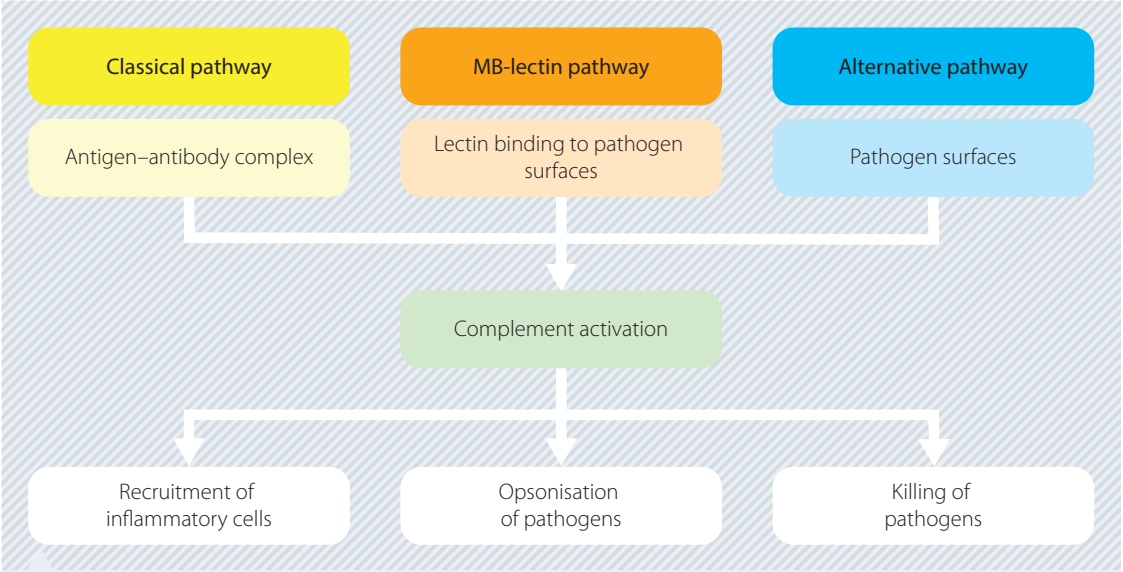


FIGURE 11.31 Monocytes are capable of transforming into three different cell types.

The complement system responds chemically to pathogens

The **complement system** is a group of around twenty soluble proteins that assist other defence mechanisms (Fig. 11.32) in destroying extracellular pathogens. These complement proteins can stimulate phagocytes to become more active, attract phagocytes to the site of the infection or destroy the membranes of the invading pathogen.



Source: Adapted from Janeway CA Jr, Travers P, Walport M, et al. The complement system and innate immunity. 'Immunobiology: The Immune System in Health and Disease'. 5th edition. New York: Garland Science; 2001. Figure 2.7 'Schematic overview of the complement cascade'

FIGURE 11.32 The role of complement in responding to pathogens

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Complement proteins are manufactured in liver cells and macrophages. These proteins circulate in the blood and are a part of the initial response to infection. During the third line of defence, some pathogens bind to proteins in the blood called antibodies (Chapter 12). Complement proteins are attracted to pathogen-antibody complexes and bind to them as well. A layer of complement proteins now coats the antigen–antibody complex. This acts as a signal for phagocytes and other lymphocytes called B cells (third line of defence) to destroy the pathogen. This enhancement of the process is called **opsonisation**. Some complement proteins can 'punch' holes in microbial cell membranes, causing the cell contents to leak out (Fig. 11.33). All these processes form part of the innate immune response to pathogens. (This will be discussed in more detail in Chapter 12.)

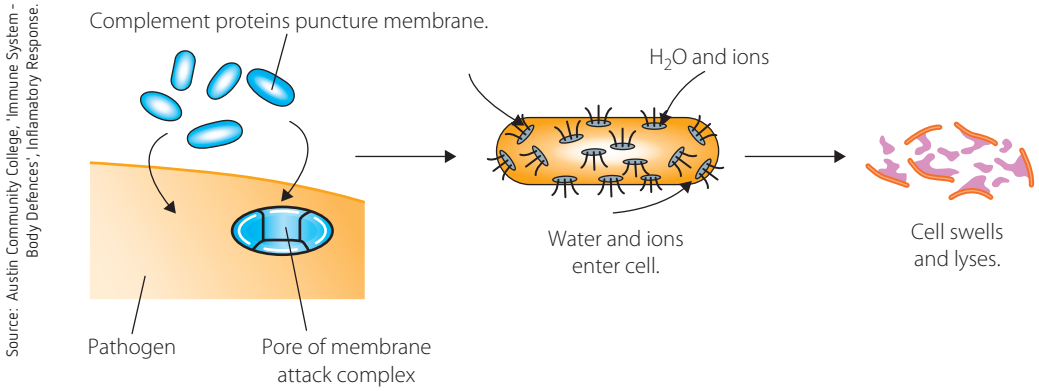

FIGURE 11.33 Complement proteins can destroy pathogens by inserting themselves into the membrane of the pathogen, creating a pore.

Fever: the role of pyrogens as a chemical response to pathogens

The hypothalamus is a part of the brain that contains a special cluster of cells responsible for regulating body temperature. It acts like a thermostat to initiate responses to keep the body temperature within a certain set range. Normal body temperature for humans is (on average) 37°C.

The body may react to pathogens by altering the hypothalamic set point of body temperature and allowing the tissue to heat up. It does this by releasing 'fever-causing' chemicals known as **pyrogens** ('fire starters') (Fig. 11.34). We experience this elevation in body temperature as a fever. The purpose of a fever is to kill or limit the growth of pathogens. It may also enhance the activity of white blood cells and thereby strengthen the response to the presence of the pathogen. The scientific name for fever is **pyrexia**. Fever is usually accompanied by sweating, chills, muscle aches and general weakness.

Regulation of body temperature is dealt with in Chapter 14

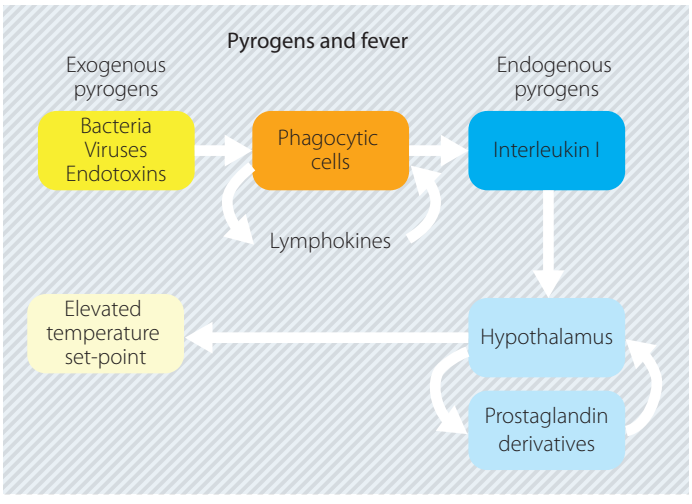

FIGURE 11.34 Pyrogens are produced by both pathogens and phagocytes, acting to raise the hypothalamic set point.

INVESTIGATION 11.4

The effect of temperature on the viability of pathogens

Critical and creative thinking

Information and communication technology capability

Raising the temperature of the body is one response strategy to slow down or destroy pathogens. Fever is a complex series of interactions between the site of infection and the brain. Chemicals called pyrogens mediate fever. In this activity, you will design a primary investigation to analyse the effect of increased temperature on pathogen growth rates.

Growing pathogens at or near body temperature is a high-risk activity and therefore you will simply design an experiment that could be tested in a facility with the appropriate equipment and protocols to protect you from biological hazards.

AIM

To design and plan an investigation in order to obtain primary and secondary data on the effect of increased temperature on the growth rate of a named pathogen

HYPOTHESIS

Write a suitable hypothesis for your investigation.

METHOD

1. Choose a named pathogen (choose from bacteria, fungi, viruses, protozoa).
2. Write a method to test your hypothesis.
3. You may need to refer to online and printed secondary resources to gather information regarding how these pathogens are cultured in a laboratory.
4. Think about how you will be able to expose the pathogens to a range of temperatures.

RESULTS

Design a results table to record your observations.

DISCUSSION

Analyse the possible links between this experiment and the role of fever in the body in response to the presence of pathogens.

Although a temporary (around 2–3 days) and mild fever is a normal response to pathogen invasion, very high fever for a prolonged period is a cue to seek further medical advice, as it may be a sign of significant illness and elevated blood temperature in the brain can cause seizures. An unexplained fever in a child, especially younger than three months, is cause for investigation by a doctor, particularly if the child is listless, vomiting or unresponsive to eye contact.

Worksheet
Link between cytokines, inflammation and non-infectious diseases

Cytokines

Cytokines are chemical messengers that are produced during an infection and they promote the development and differentiation of T and B lymphocytes for the third line of defence. One example is **interleukin** (IL). These chemicals form a link between the innate and adaptive immune systems. A burst of cytokines occurs as infected cells signal to nearby uninfected cells to also release cytokines. **Interferons** are another example of cytokines. They act as a chemical signal to viruses to stop replicating. They do this by signalling to uninfected cells to destroy RNA and reduce protein

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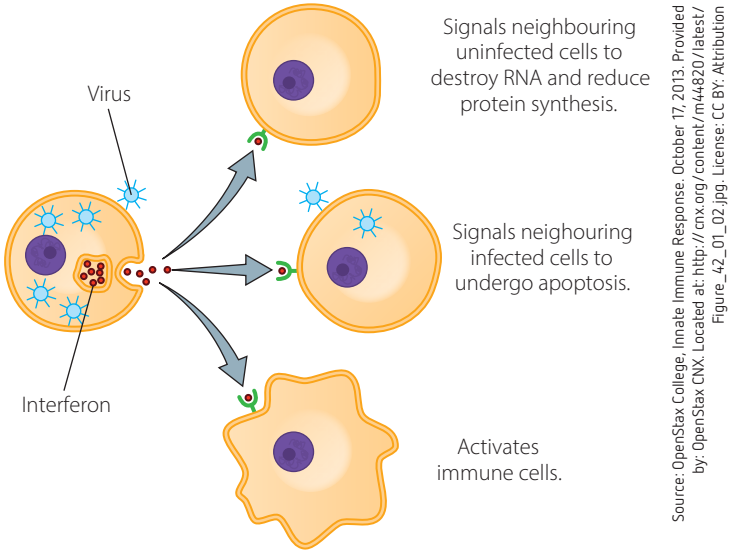
synthesis (Fig. 11.35). Infected cells are also signalled to undergo apoptosis (programmed cell death). Cytokines play a role in the feelings of lethargy, muscle pain and nausea that you experience if you have an infection. The implication of this is that the animal isolates itself and rests, and is therefore prevented from spreading the infection to other animals.



Source: OpenStax College, Innate Immune Response. October 17, 2013. Provided by: OpenStax CNX. Located at: http://cnx.org/content/m44820/latest/Figure_42_01_02.jpg. License: CC BY: Attribution

FIGURE 11.35
Cytokines are chemical signals released by cells in response to pathogens.

KEY CONCEPTS

- Inflammation is a response to the presence of pathogens in the tissues.
- Specialised cells and tissues use a series of chemical signals to identify and destroy pathogens.
- The signs of inflammation are mediated by chemical signals and include redness, heat, swelling, pain and loss of function.
- Histamines, bradykinin, serotonin and prostaglandins are the chemical mediators of the signs of inflammation.
- Chemical signals include the complement system, cytokines and PAMPs.

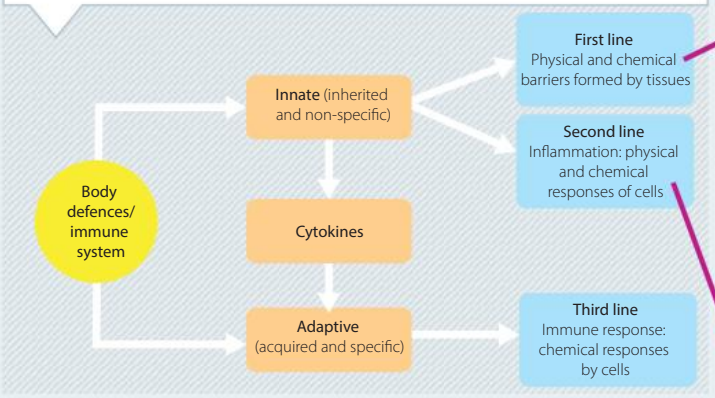
CHECK YOUR UNDERSTANDING
11.2e

1. How do the five cardinal signs of inflammation relate to a chemical response from the tissues and cells of an infected organism?
2. Describe how the process of phagocytosis destroys pathogens in response to a chemical signal.
3. A blood smear from a young person with a fever is examined under a light microscope. The pathologist sees elevated numbers of circulating neutrophils. Many of them appear toxic. Explain the presence of these cells in the young person's blood. What process is likely to be occurring?
4. Explain the presence of monocytes in an infected tissue sample, if monocytes circulate in the blood.
5. Explain the presence of complement proteins at the site of an infection.
6. Analyse the role of cytokines in the destruction of pathogens.

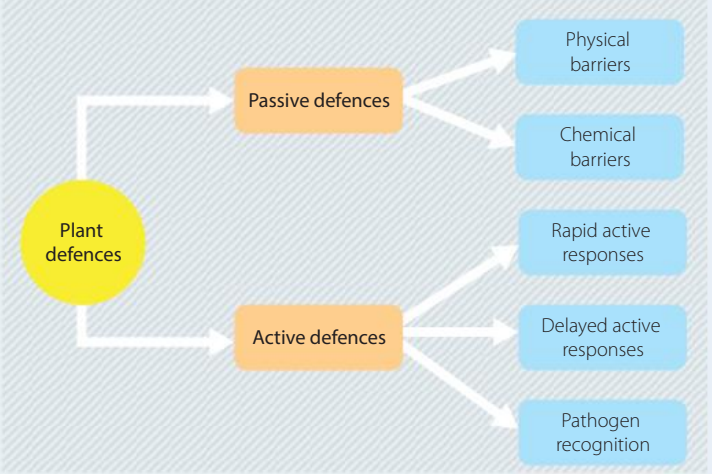
11 CHAPTER SUMMARY

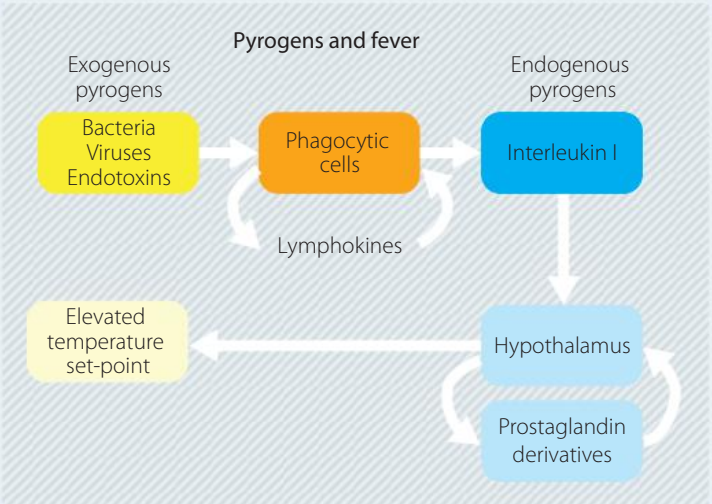
Responses to pathogens: How does a plant or animal respond to infection?

ORGANISATIONAL LEVELS OF THE BODY'S DEFENCES



RESPONSES BY PLANTS AGAINST PATHOGENS





First line of defence

Second line of defence

Inflammation

A chemical response that helps wound repair and leads to pathogen destruction. The five cardinal signs of the inflammatory response: dolor (pain), calor (heat), rubor (redness), tumor (swelling), function laesa (loss of function).

Phagocytosis

The process by which phagocytes change shape and completely enclose a foreign particle, releasing enzymes to destroy it.

The main types of phagocytes: neutrophils, monocytes/macrophages, dendritic cells, natural killer cells.

Fever

Pyrogens are fever-causing chemical. The purpose of a fever is to kill or limit the growth of pathogens

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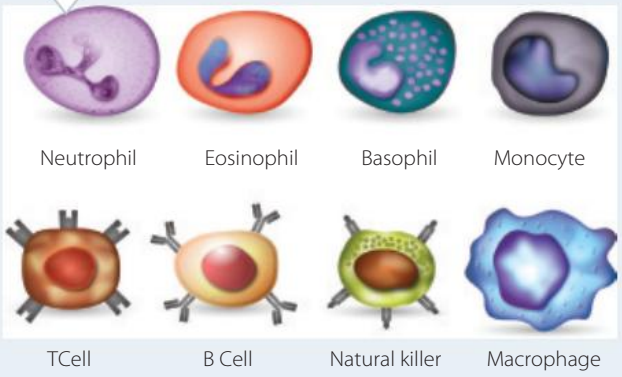
TABLE 11.3 Cells involved in chemical and physical responses to infection by pathogens (lymphocytes are not included)

CELL TYPE	CHARACTERISTICS	LOCATION
Mast cell	• Blood vessel dilation • Release of heparin and histamines • Recruitment of neutrophils and macrophages • Also involved in allergic reactions	Connective tissues and mucous membranes
Macrophage	• Phagocytosis of pathogens and cancer cells • Antigen-presenting cell	Migrates from blood vessels into tissues
Natural killer cell	• Kills tumour cells and virus-infected cells	Circulates in blood and migrates into tissues
Dendritic cell	• Antigen-presenting cell • Triggers adaptive immune response	Epithelial tissues, including skin, lung and tissues of the digestive tract. Migrates to lymph nodes upon activation
Monocyte	• Differentiates into phagocytic cells such as dendritic cells and macrophages	Stored in spleen. Moves to infected tissues through blood vessels
Neutrophil	• Most common white blood cell at site of trauma or infection • Releases toxins that kill or inhibit bacteria and fungi • Recruits other immune cells to the site of infection	Migrates from blood vessels into tissues
Basophil	• Defence against parasites • Releases histamines that cause inflammation • Responsible for some allergic reactions	Circulates in blood and migrates to tissues
Eosinophil	• Releases toxins that kill bacteria and parasites	Circulates in blood and migrates to tissues

Source: adapted from Lumen Learning Canada 2017, Boundless Biology, 'Innate immune response', https://courses.lumenlearning.com/boundless-biology/chapter/innate-immune-response/

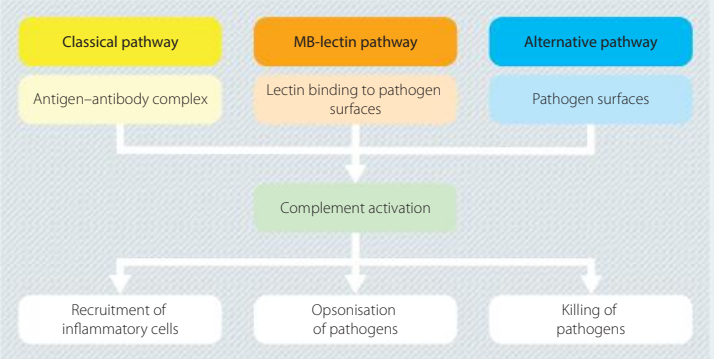
An *antigen* is any molecule that the body recognises as foreign and that triggers an immune response.

CELLS INVOLVED IN CHEMICAL AND PHYSICAL RESPONSES TO INFECTION BY PATHOGENS



COMPLEMENT ACTIVATION

The complement system is a group of proteins that assist other defence mechanisms in destroying extracellular pathogens. They stimulate phagocytes to become more active, attract phagocytes to the site of infection or destroy the membranes of the invading pathogen.



11 CHAPTER REVIEW QUESTIONS

Review quiz

1. Explain why both plant and pathogen factors are important in the development of disease in a plant.
2. Describe the ways in which plants use barriers as a passive defence against pathogens.
3. PAMPs are chemicals secreted by bacteria that alert a plant to the presence of that pathogen. How do plants respond to the presence of PAMPs?
4. The active defences available to a plant are initiated when it is at grave risk of harm. Outline the three main strategies that plants use when passive barriers are breached.
5. Compare the responses of plants and animals to the presence of pathogens in terms of physical and chemical strategies used when threatened with an infection.
6. Justify the practice of carefully disposing of plant material after an experiment.
7. Describe the organisation of the animal immune system into 'lines of defence'.
8. Contrast the innate and adaptive immune systems.
9. Outline the way in which the cells of the immune system distinguish foreign molecules from 'self'.
10. Describe the usefulness of the lymphatic system in terms of immune surveillance.
11. The loss of microflora from the skin or gut can lead to ill health. Explain why microflora are needed for good health.
12. Describe the role of the skin as a barrier to infection.
13. Explain the various mechanisms by which mucous membranes prohibit entry of pathogens.
14. Explain why granulomas sometimes form in response to the presence of a pathogen. Use a specific example of a disease in which this occurs.
15. Vomiting and diarrhoea are very unpleasant, but are extremely important strategies against infection. Outline the role of these processes in the response of animal tissues to an infection.
16. Discuss some of the strategies used by an animal's body to ensure one-way flow of materials. How does this reduce the likelihood of infections?
17. Outline the role of urine, sweat, tears and gastric juice as chemical barriers to infection.
18. In animals, a fever often develops in response to an infection. What chemical signals are involved in this process?
19. Outline the role played by the complement system in response to the presence of pathogens.

Exam preparation

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12 Immunity

INQUIRY QUESTION

How does the human immune system respond to exposure to a pathogen?

- investigate and model the innate and adaptive immune systems in the human body (ACSBL119)
- explain how the immune system responds after primary exposure to a pathogen, including innate and acquired immunity

Getty Images / Steven Debenport

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Worksheet
Immunity

Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
12.1 A secondary source investigation into chemical signalling between the innate and adaptive immune systems

12.2 A practical investigation using the light microscope to examine the cells and tissues of the innate and adaptive immune systems

12.3 Changes in the blood during infectious disease

12.4 A practical investigation to model the responses of the innate and adaptive immune systems

Worksheets
- Immunity
- Antibodies
- Chemical mediators and receptors of the immune response
- Cells of the adaptive immune system

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

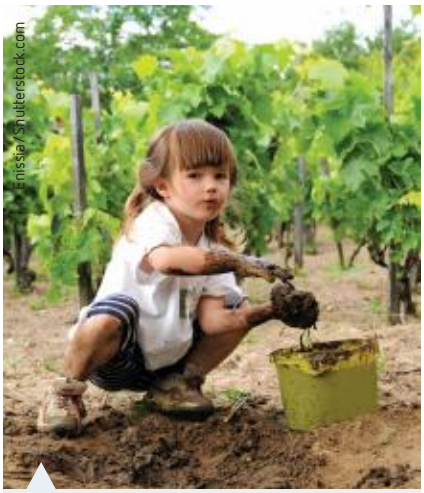
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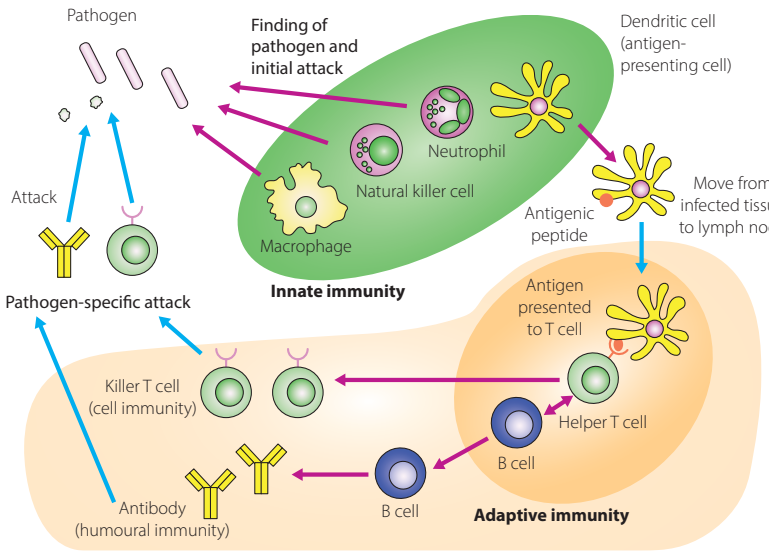
FIGURE 12.1 We are surrounded by pathogens from a very early age.

We are surrounded by a sea of pathogens. Yet, for the most part, we do not spend our lives consistently infected with disease. This is due to the integrated actions of the innate and adaptive immune systems in the human body, which work together to recognise and eliminate potential invaders. Scientists think the elements of our immune system evolved around 500 million years ago, in jawed vertebrates called gnathostomes.

12.1 The human immune system

The human immune system is able to recognise patterns, and to alert other cells and tissues when the patterns are perceived as 'non-self'. It sets into motion a chain of events aimed at eliminating infection. Many of the mechanisms by which the innate and adaptive immune systems communicate with each other are still not well understood. However, advances in molecular biology are slowly revealing the complex ways in which these systems communicate to keep us safe (Fig. 12.2). Host defence systems such as the innate and adaptive immune systems arose from the harmful effects



Source: Royal Society Publishing, Philosophical Transactions of the Royal Society B, 'Innate immunity and adjuvants', by Shizuo Akira, 5 Sep 2011, Figure 1. (CC BY 4.0) (https://creativecommons.org/licenses/by/4.0/)

FIGURE 12.2 The innate and adaptive immune systems communicate and work together to destroy pathogens. The area of the diagram shaded green shows the innate response and the buff-coloured area shows the adaptive response.

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of pathogens acting as a selection pressure on organisms. The unique arrangement and diversity of cellular responses in these two systems is found only in vertebrates with jaws.

As discussed in Chapter 11, the innate immune system provides a first and second line of defence against pathogens, through such means as:

- physical barriers (e.g. skin)
- body fluids (e.g. urine, mucus)
- non-specific cellular responses (e.g. phagocytosis)
- non-specific biochemical responses (e.g. complement activation, endogenous pyrogens).

The *innate immune response* is genetically pre-programmed. With repeated exposure to the same pathogen, the body responds in exactly the same way. This is present from birth and does not alter, except in circumstances of poor health due to other problems. It involves no 'learning 'or 'memory formation' by the body.

The *adaptive immune response* is the next line of defence. It swings into action when the innate immune response fails to clear the pathogen from the body. It is often referred to as the third line of defence, or acquired immunity.

Adaptive immune response

There are two 'arms' of the adaptive immune response.

- The **humoral response** is effective against pathogens in body fluids.
- The **cell-mediated response** is effective against intracellular pathogens.

The cells responsible for generating the adaptive immune response are known as B and T lymphocytes (or B and T cells).

- Some B lymphocytes (B cells) develop into plasma cells, which produce **antibodies** (also called **immunoglobulins**) against the pathogen. Antibodies are protein molecules with a specific molecular structure that helps them to recognise and bind to specific pathogens. This is also known as the *humoral response*. The word 'humor' was once used to describe the main fluids of the body. The humoral response occurs in the blood and tissue fluids of the body.
- T lymphocytes (T cells) transform into cytotoxic T cells (also known as 'killer' T cells) and seek out infected body cells, binding to them and destroying them. This is also known as the *cell-mediated response*.

Figure 12.4 shows a possible scenario and pathway for a pathogen as it breaches the body's defences.

The adaptive immune response to pathogens is different from the first two lines of defence, in the following ways.

- It is specific.
- It involves a great diversity of possible responses.
- It has memory.
- It is capable of self-tolerance.

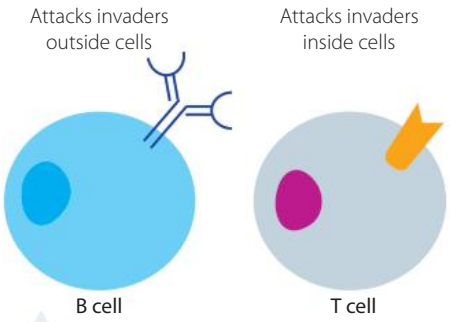


FIGURE 12.3 B and T lymphocytes target different types of antigens.

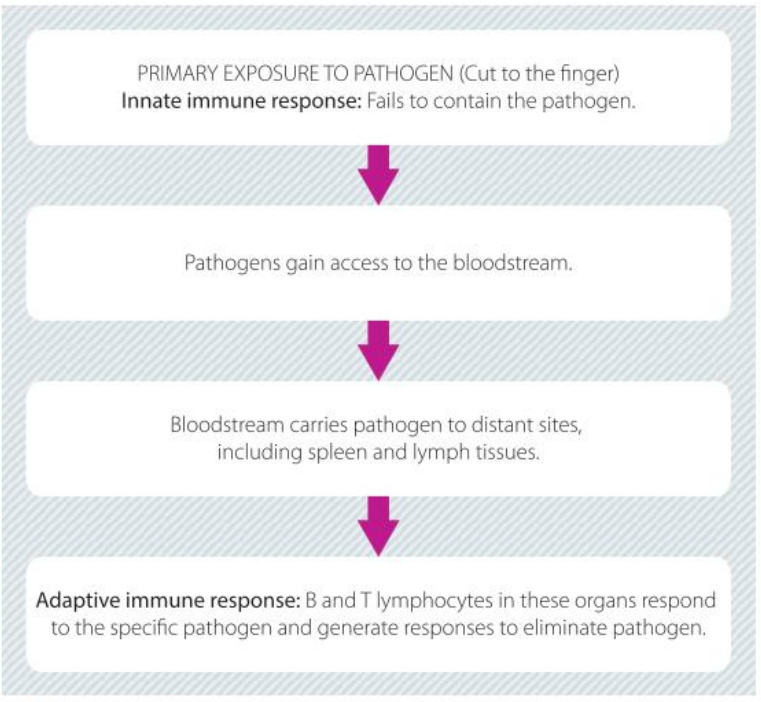


FIGURE 12.4 Initiation of the innate and adaptive immune systems

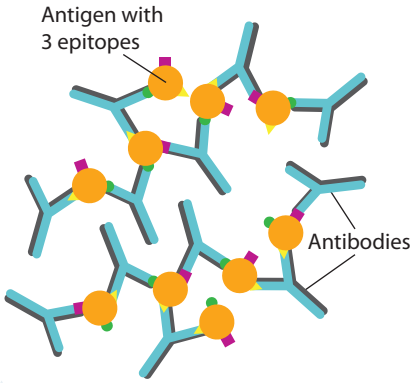
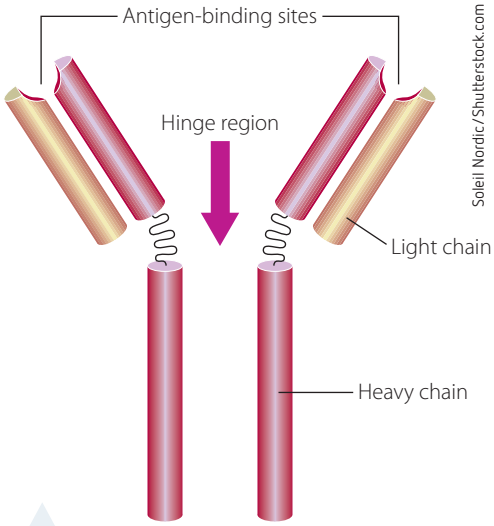


FIGURE 12.5 An antigen–antibody complex



Soleil Nordic/Shutterstock.com

FIGURE 12.6 General structure of an antibody

The specific response

Antigens are components of bacteria, viruses, fungi, worms, tumour cells and other non-cellular materials. Antigens are generally molecules made of protein or polysaccharides found on cell membranes and in viral coats. Antigen molecules have regions known as **epitopes** that can be recognised by specific lymphocytes in the immune system. Particular lymphocytes recognise the specific chemistry of each epitope and can then target these antigens. B and T lymphocytes have complementary antigen binding sites, known as *antigen receptors,* on their cell membranes. Epitopes may be bound by cell receptors on B and T lymphocytes or by free antibodies in the extracellular fluid (Fig. 12.5).

Antibodies are Y-shaped molecules that consist of four chains of protein – two larger heavy chains and two smaller light chains (Fig. 12.6). There are five types of heavy chains, which gives the five classes of antibodies: IgA, IgE, IgG, IgM and IgD. A specific region of the molecule serves as a binding site for antigens via the epitopes.

An antibody molecule has two binding sites. Each antigen-binding site is specific for a particular antigen. Pathogens may have several different epitopes on their surface that will bind to different antibodies. When antibodies and antigens bind, the resulting molecule is known as an antigen–antibody complex.

B cell membrane antigen receptors have the same affinity for a specific pathogen as the antibodies that this cell will secrete once it is transformed into a plasma cell. T cell receptors are surface proteins but their structure is unlike that of antibodies. They are not secreted from T cells but remain bound to the cell membrane.

Diversity of pathogens

T and B cells have thousands of antigen receptors on their cell membranes. For a single T or B cell, however, each of the receptors is capable of recognising only one of the millions of possible antigens in the world. However, each B and T cell is different. Therefore, even though each B or T cell shows specificity, there are millions of combinations of receptor types available to the human immune system. The combinations are determined by a person's genes and so are programmed from birth. Your adaptive immune system is primed and ready to respond to millions of antigens it has not yet seen and may never see.

The clonal selection theory states that all the B cells for all the possible antigens are *already present* in very small amounts in the immune system at birth. This is an astounding concept when you consider the number and variety of things in the world that the body could perceive as 'non-self'. Antigens are recognised as 'non-self'. When an antigen is present in the body, the B cell that is specific for that antigen is activated, and then cloned. Once the antigen is destroyed, these cloned B cells remain, ready for the next time this specific antigen presents itself to the body – they become memory cells. Think of an army that has been shown an image of the enemy, and now lies in wait for that enemy to approach. It is primed and ready to respond. It has 'adapted'.

This explains why the adaptive immune response is so specific. If a particular cold virus enters your body, only the B and T cells that recognise that pathogen will respond to it.

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Primary immune response

When the adaptive immune system is first exposed to a pathogen, the response by the B and T lymphocytes is referred to as a **primary immune response** (primary refers to 'first'). Antibody-producing plasma cells and cytotoxic T cells are produced, and these work to clear the infection. The body stores these pre-made cells for the next time it encounters the same pathogen. The person will possibly now be immune to infection by this pathogen. This immunity is therefore acquired or adaptive.

MHC molecules are explained on page 405.

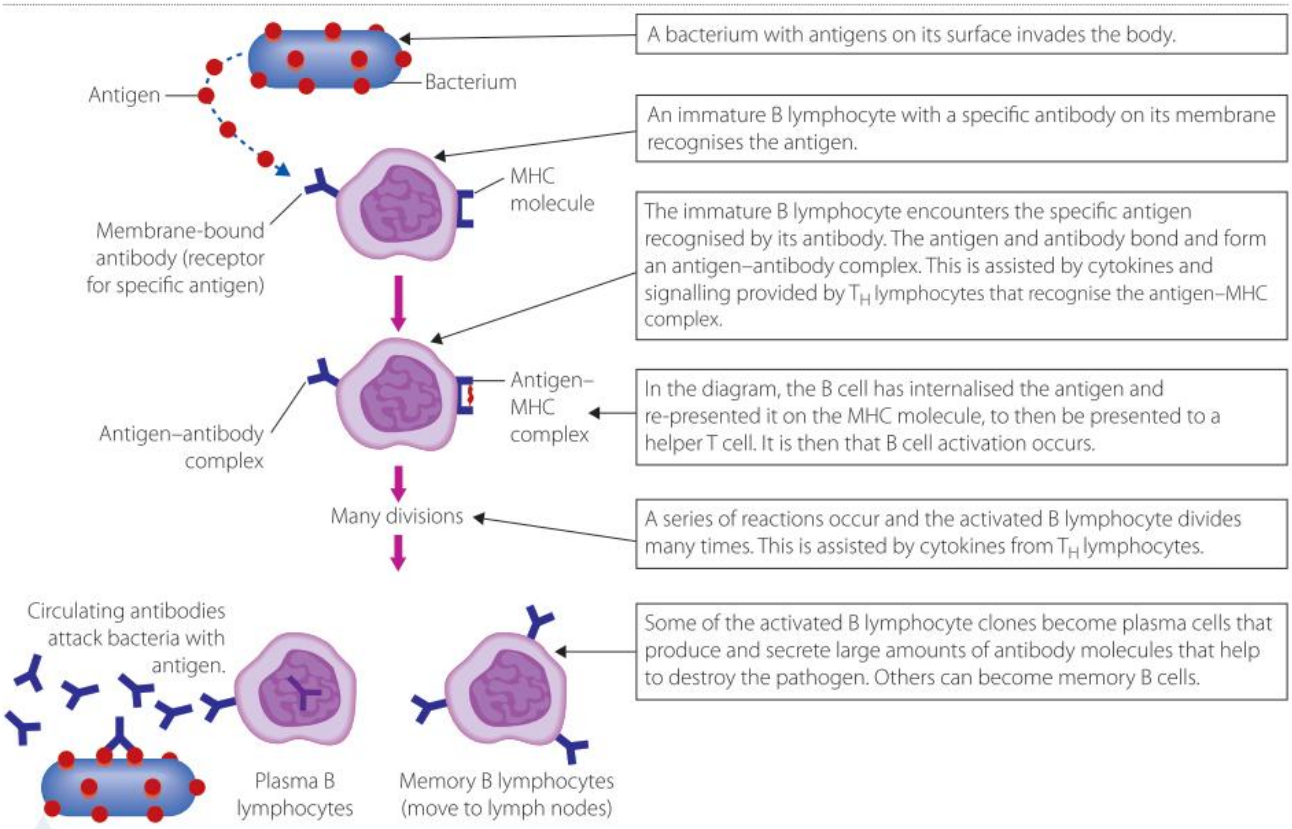

FIGURE 12.7 B cells for all possible antigens pre-exist in the body and are cloned when exposed to that specific pathogen. This is how 'memory' of an antigen is formed (MHC: major histocompatiblity complex; T_H: helper T cell).

This response is fairly fast but is short lived. The total number of antibodies produced in this initial response is less than for subsequent infections. When the adaptive immune system is exposed to the same pathogen on subsequent occasions, there is a more rapid response and the production of antibodies is much greater. This is also referred to as a **secondary immune response**. This is the reason that booster vaccinations are often recommended for various diseases. The secondary immune response comes about because of the existence of memory T and B cells that were produced in the first infection with the pathogen.

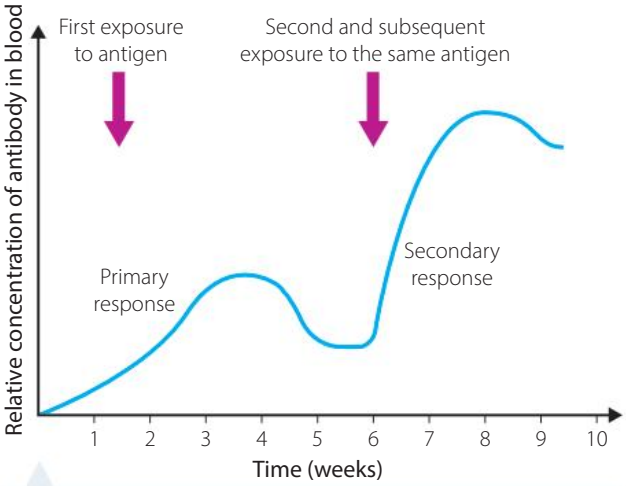

FIGURE 12.8 The primary and secondary immune responses

Do no harm

Fortunately, despite the fact that a person's immune system is genetically pre-programmed to respond to millions of possible antigens, it does not normally react to the tissues of that person. How is this achieved?

When you go to school, you learn about the various rules and regulations of society that help everyone live and work safely. In your school, rules and boundaries are established to keep everyone safe and provide a comfortable learning environment. It is not acceptable for individuals to turn against and harm those with whom they share a school, workplace or city. B and T cells also have a period in which they are 'in school'. They must learn not to harm the body in which they have developed. When they start to mature in the bone marrow (B cells) and thymus gland (T cells) they are examined for possible tendencies to cause harm to the body. Cells that have a genetic tendency to identify self-molecules as foreign are identified and neutralised, so they never get to mature and cause harm to the body. The only lymphocytes that remain are those that are capable of reacting to 'other' and not to 'self'. Scientists think that autoimmune diseases may occur when this process has not been fully effective.

KEY CONCEPTS

- The innate and adaptive immune systems defend an organism against invasion by pathogens.
- The innate immune system is genetically programmed and present at birth. It consists of non-specific chemical and physical barriers as well as chemical and cellular responses to pathogens.
- The adaptive immune system responds to pathogens that have evaded the innate immune system.
- The adaptive immune system responds to specific pathogens, is capable of responding to a diverse array of pathogens, remembers a primary response for future threats by the same pathogen, and tolerates the host's own cells.

CHECK YOUR UNDERSTANDING 12.1a

1. By what four general means does the innate immune system respond to pathogens?
2. When the innate immune response is ineffective, which immune response swings into action?
3. Identify the four main features of the adaptive immune response on primary exposure to a pathogen.
4. Which two cell types are responsible for the adaptive immune response? Outline the role of each of these cell types.
5. Outline the ways in which the responses of the adaptive immune system towards pathogens:
   - **a** are specific
   - **b** are diverse
   - **c** remember and tolerate 'self'.
6. Distinguish between a primary and a secondary response to pathogens by the adaptive immune system.
7. Use your understanding of the humoral immune response to justify the shape of the curve in Figure 12.8.

The humoral response to pathogens in body fluids

After primary exposure to a pathogen that is outside the body cells (extracellular pathogen), the adaptive immune response has strategies to target and inactivate the pathogen – this is the humoral immune response.

B lymphocytes are the cells primarily responsible for the adaptive immune response outside cells. Mature B lymphocytes are stored in lymph nodes and peripheral lymphoid tissues, and circulate in the blood. Lymph nodes are special glands located at key points around the body where tissue fluids drain from major sites (Fig. 11.13).

Helper T cells release substances such as cytokines that are involved in the activation of B cells. Two adaptive responses follow (Fig. 12.9).

1. The B cell multiplies, making many copies of itself with the same specificity as the original B cell.
2. These B cells differentiate into two possible cell types:
   - plasma cells – short-lived antibody 'factories' or effector cells (secrete up to 2000 molecules/second)
   - memory cells – long-lived and stored for subsequent infections.

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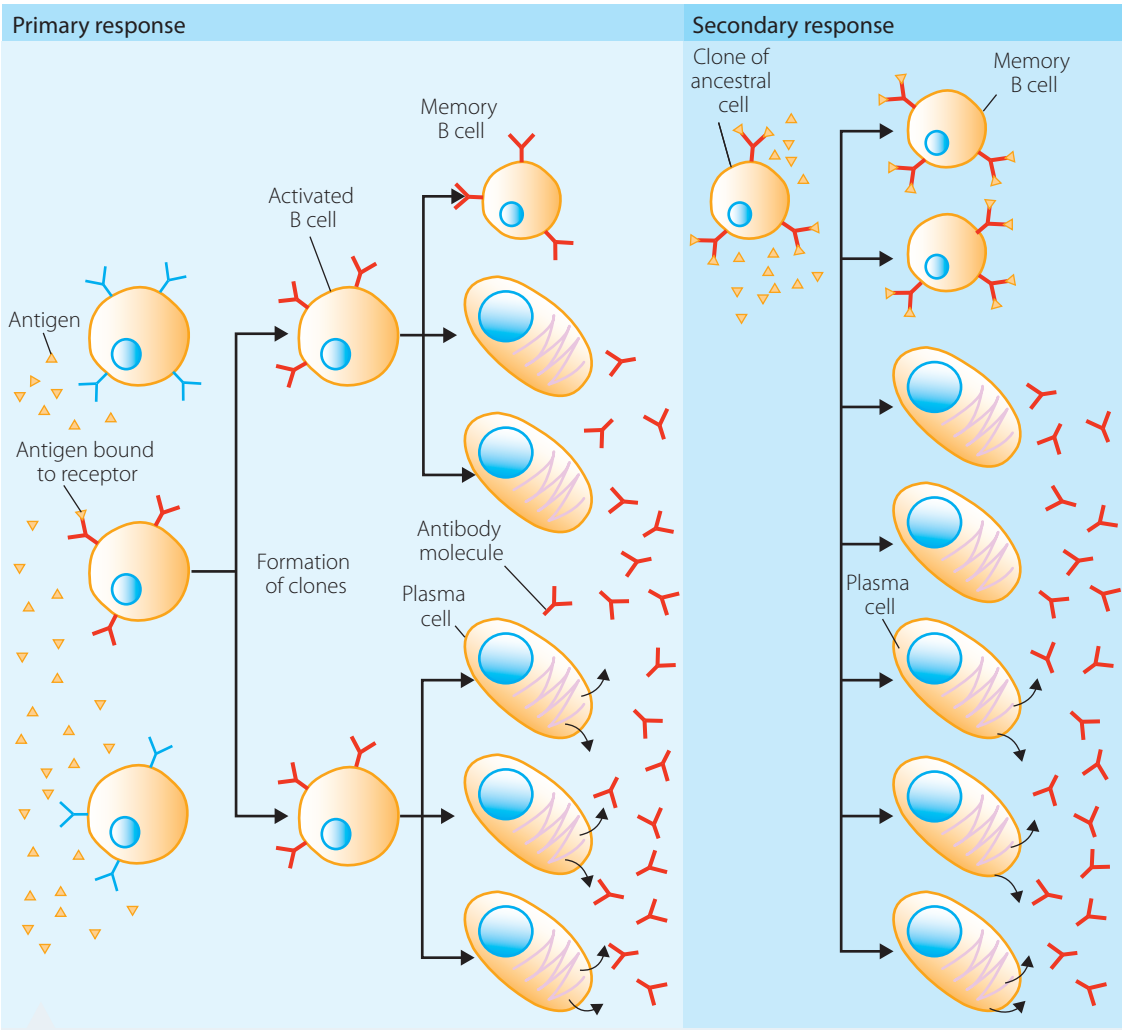
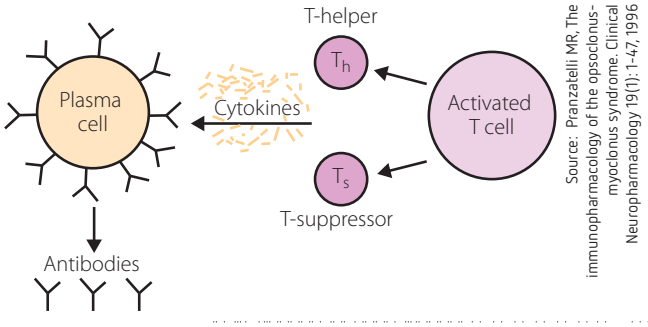


FIGURE 12.9 The two possible fates of activated B lymphocytes

B lymphocytes

When protein-containing antigens are present, specific T cells, called helper T cells, are activated when they bind to the antigen. The T cells release special molecules known as **cytokines**. Examples of cytokines include the molecule interleukin-2 (IL-2). When B lymphocytes are exposed to IL-2, they proliferate and differentiate into memory cells and plasma cells (Fig. 12.10).

FIGURE 12.10
Activated T cells assist in the activation and transformation of B cells into plasma cells.



Not all antigens promote the production of memory cells. Some bacteria have antigens composed of long polysaccharide chains (not protein), which bind directly to B lymphocytes, causing them to multiply and differentiate into plasma cells. However, no memory cells are made in this process. These kinds of antigens are known as *thymus-independent antigens*.

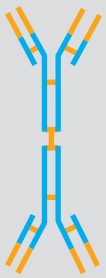
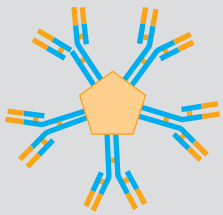
Antibodies

Worksheet
Antibodies

Antibodies are molecules produced by plasma cells in response to exposure from a specific pathogen. However, not all antibodies are the same, and nor are they found in equal concentrations around the body.

There are five classes of antibodies, also known as immunoglobulins (Ig). Table 12.1 summarises their features.

TABLE 12.1 The five classes of antibodies (immunoglobulins)

NAME	PROPERTIES	STRUCTURE
IgA	Found in mucus, saliva, tears and breast milk. Protects against pathogens.	
IgD	Part of the B cell receptor. Activates basophils and mast cells.	
IgE	Protects against parasitic worms. Responsible for allergic reactions.	
IgG	Secreted by plasma cells in the blood. Able to cross the placenta into the foetus.	
IgM	May be attached to the surface of a B cell or secreted into the blood. Responsible for early stages of immunity.	

(Source: Molnar C. & Gair J. 2013, 'Concepts of biology', 1st Canadian edition, Table 23.23, Gunness, PC Faculty Pressbooks Sites, https://pressbooks.bccampus.ca/conceptsofbiologygunness/chapter/23-3-antibodies/)

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The different classes of antibodies act in different ways to neutralise a pathogen. There are five strategies that antibodies can use to ensure that a pathogen is stopped from harming the host. The first two ways are common to all antibodies; the final three ways are special strategies used by specific antibody classes.

1 Neutralisation – antibodies bind to and coat pathogens, blocking their activity (Fig. 12.11a).
2 Agglutination – neutralised pathogens clump together and are surrounded by thousands of antibodies (Fig. 12.11b).
3 Precipitation of dissolved antigens.
4 Activation of the complement system, leading to lysis of infected cells (Fig. 12.11c).
5 Opsonisation – enhanced phagocytosis by natural killer cells (NK cells) (Fig. 12.11d).

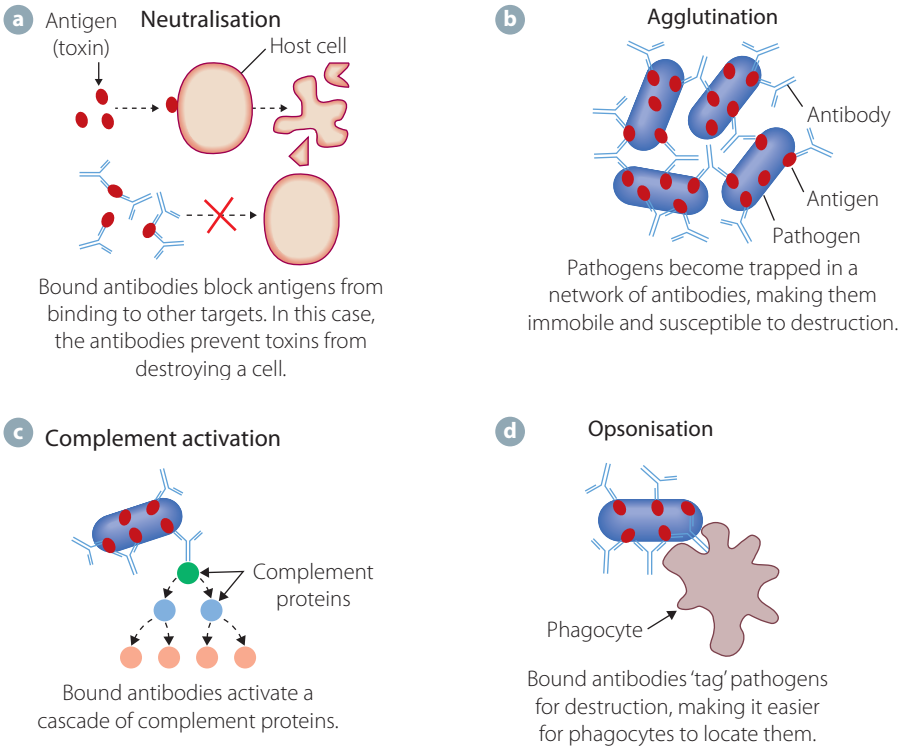

FIGURE 12.11 Four ways in which antibodies destroy pathogens: **a** neutralisation; **b** agglutination; **c** complement activation and **d** opsonisation

KEY CONCEPTS

- The humoral immune system is a branch of the adaptive immune response that targets pathogens in body fluids.
- The main type of cell involved in the humoral immune system is the B lymphocyte, which develops and matures in the bone marrow.
- Mature B lymphocytes are stored in lymph nodes and circulate in the blood.
- B cells respond to pathogens by differentiating into memory cells and antibody-producing plasma cells.
- Antibodies deal with pathogens through neutralisation, agglutination, precipitation, complement activation and opsonisation of phagocytes.

CHECK YOUR UNDERSTANDING 12.1b

1. Justify the need for the immune system to take a different approach to the elimination of extracellular versus intracellular pathogens.
2. Describe the two possible pathways that B cells may take upon exposure to an extracellular pathogen.
3. Explain the ways in which antibodies can respond to the presence of an extracellular pathogen.
4. What advantages does an antibody-mediated response give to a host organism over life forms that rely on the innate immune system alone?
5. Using your knowledge of the chemical nature of antibodies, infer the possible effects of a protein deficiency in a human due to disease or malnutrition.

The cell-mediated response to intracellular pathogens

The adaptive immune system has another branch, responsible for the elimination of pathogens located inside host cells (intracellular pathogens). Because antibodies (which function in the humoral response) have no direct access to these pathogens, another strategy is required to ensure the pathogen is destroyed. This is known as *cell-mediated immunity*, where special types of T lymphocytes target and destroy the entire infected host cell, along with the pathogens inside them. The process of cell-mediated immunity relies on the action of T lymphocytes.

T lymphocytes make direct contact with infected cells via special receptors, and can also identify tumour cells and destroy them. This is why their particular response is known as the cell-mediated immune response (Fig. 12.12).

T lymphocytes control cell-mediated immunity, which is effective in defending the body against:

- protozoa, bacteria and viruses that are inside the host's body cells
- macroparasites such as fungi, flatworms and roundworms (despite the fact that they are not intracellular parasites)
- cancer cells and transplanted tissues (e.g. in a person who has had a kidney or lung transplant).

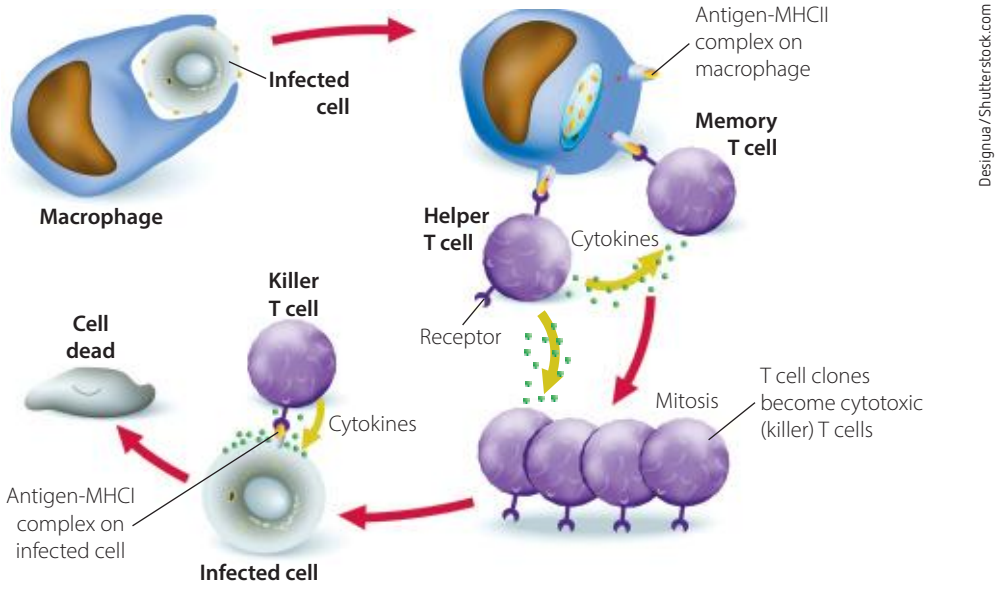

FIGURE 12.12 The cell-mediated immune response

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T cells and MHC molecules

In the previous section, you learned that antibodies bind to the epitopes of pathogens in their natural form. However, T cells recognise fragments of antigens only. These may be antigens that have been party digested by macrophages (Fig. 12.13) or antigens partly digested by invaded cells. The partly digested antigens are carried to and displayed on the cell surface by special protein molecules called **major histocompatibility complex molecules** (MHC).

MHC molecules are the 'self-identity' molecules of organisms. There are over a hundred genes that code for an individual's MHC marker molecules. Each person has a different set of MHC marker molecules on their cells. These molecules have a binding site for a specific antigen. There are two classes of MHC molecules:

- MHCI – in every nucleated cell in vertebrates (including platelets, but excluding red blood cells, which do not have a nucleus). Infected cells represent a fragment of antigen bound to an MHCI molecule.
- MHCII – in antigen-presenting cells such as macrophages, dendritic cells and activated B cells. Macrophages present a fragment of the antigen bound to an MHC II molecule. This is termed an antigen-MHCII complex (Fig. 12.12).

T lymphocytes are produced in the **primary lymphoid tissue** of the bone marrow and mature in the thymus gland, which is situated in the thoracic (chest) cavity. After they mature, the T cells are released into the blood, and migrate to the spleen, tonsils, **Peyer's patches** of the intestines and lymph nodes (Fig. 11.13, page 374). The four main types of T cells (summarised in Fig. 12.13) are described below.

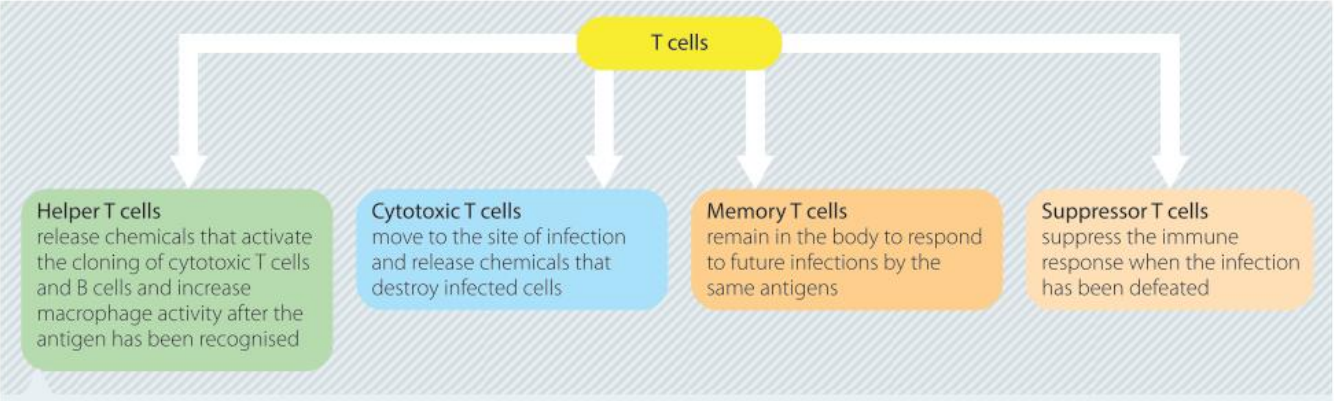

FIGURE 12.13 Types of T cells and their roles

Helper T cells

Helper T cells have a surface protein receptor that recognises the MHCII molecules bound to an antigen fragment on the surface of phagocytes ('antigen-presenting cells') (Fig. 12.12). The helper T cells are activated to secrete cytokines, such as IL-2 (interleukin-2), which activate both the cell-mediated and humoral responses. IL-2 activates cytotoxic T cells and B cells that are specific for this antigen. In this way, helper T cells are the 'bridge' between the two arms of the adaptive immune response. Other cytokines that stimulate the activity of macrophages are also released by the helper T cells.

Cytotoxic T cells (Tc cells)

Cytotoxic T cells are activated by helper T cells and respond by producing many copies (clones) of themselves. In this way, they form an 'army' of identical killer cells that move to the site of infection, bind with the infected cells and release chemicals (cytokines) that destroy the infected cell. Cytotoxic T cells use their surface receptor to recognise the MHCI–antigen complex (Fig. 12.12) typical of cells

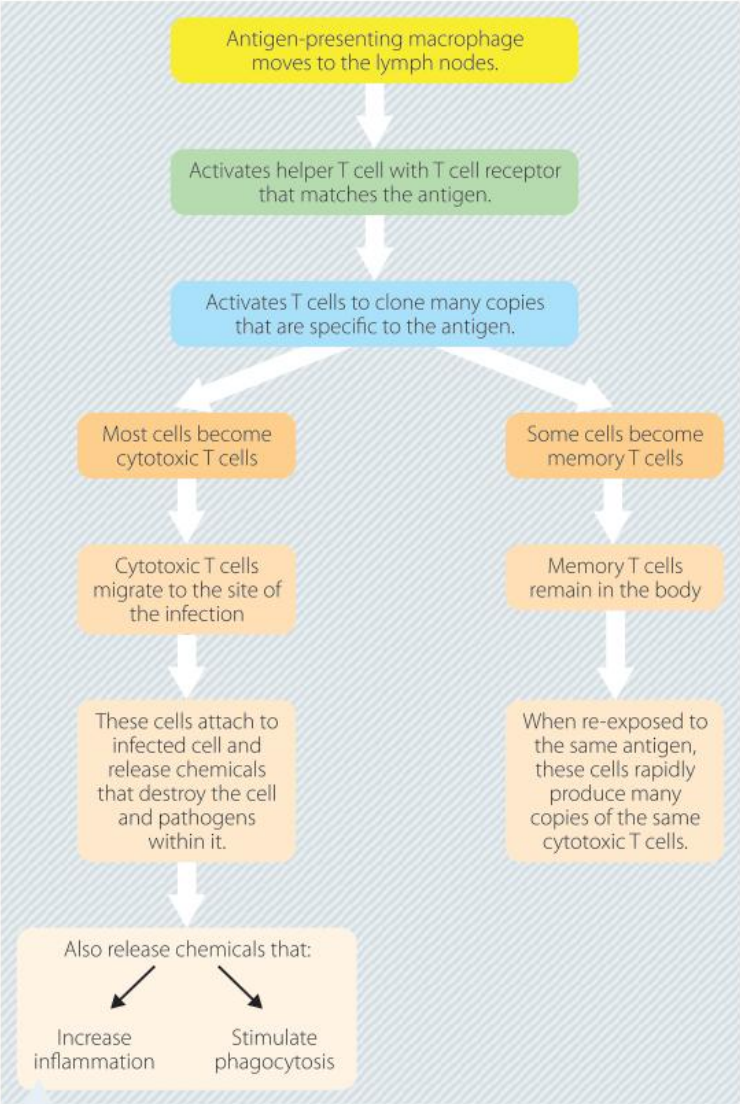

FIGURE 12.14 Steps in the process of cell-mediated immunity

that have been infected by a virus. Through a series of reactions, the cytotoxic T cell is able to then kill the infected cell. This involves the production of cytokines, which cause lysis of the infected cell.

Suppressor T cells and memory T cells

Suppressor T cells (Ts cells) are responsible for stopping the adaptive response when the infection has been defeated. **Memory cells** – all T cells make clones of themselves that remain in the body to respond to future exposure from pathogens.

Summary of cell mediated immunity

The process of cell-mediated immunity is carried out by T cells. (See the flow chart in Fig 12.14, the summary below and the visual representation in Fig 12.16.)

1. Foreign material is engulfed by macrophages, which then display the antigen attached to their MHCII molecules.
2. The antigen-presenting macrophages move to the lymph nodes, where they are inspected by helper T cells that have the T cell receptor that corresponds to the antigen being presented.
3. These helper T cells then activate the cloning of millions of cytotoxic T cells and memory T cells that are specific for this antigen.
4. The cytotoxic T cells leave the lymph nodes and migrate to the site of the infection, where their antigen receptors bind with the antigen displayed on the infected cell.
5. These T cells then release chemicals that destroy the cell and any pathogens within it.
6. These chemicals also increase the inflammation and attract more macrophages, which carry out phagocytosis to help destroy the pathogens and clear up any debris.
7. Some of the cytotoxic T cells produce a chemical, called interferon, which protects the healthy cells around an infected cell from viral invasion.
8. Once the infection has been defeated, the suppressor T cells release other chemicals to stop the production and action of the cytotoxic T cells.
9. The memory T cells that are produced at the time and are specific to that particular antigen remain in the body, in the lymph nodes. On re-exposure to the same antigen-containing pathogen, they cause the rapid production of more of the same cytotoxic T cells. This prevents the body from developing the symptoms of the disease again.

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KEY CONCEPTS

- The cell-mediated immune system is a branch of the adaptive immune response that targets pathogens inside infected cells as well as tumour cells.
- The main cell involved is the T lymphocyte, which develops in the bone marrow and matures in the thymus.
- Mature T lymphocytes are present in the blood, spleen, tonsils, Peyer's patches and lymph nodes.
- There are four main types of T lymphocytes.
  - Helper T cells activate the cloning of cytotoxic T cells as well as B cells.
  - Cytotoxic T cells bind with pathogen-infected cells and destroy them.
  - Memory T cells remain in the body to respond to future infections.
  - Suppressor T cells suppress the response once pathogens have been eliminated.

CHECK YOUR UNDERSTANDING 12.1c

1. Evaluate the importance of the placement of lymph node sites around the body.
2. Why is an alternative strategy to antibody production needed for the elimination of intracellular pathogens?
3. Which lymphocyte provides a common link between the two arms of the adaptive immune system? Describe its role in responses to intracellular and extracellular pathogens.
4. Which types of pathogens/cells are most likely to trigger a cell-mediated response?
5. Outline the role of MHC molecules in the activity of cytotoxic T cells.
6. Suggest the consequences of a lack of activity by suppressor T cells. Use online resources to investigate whether there are any known diseases where this is a problem.
7. Glucocorticoids are a class of drugs that are used to suppress various allergic and inflammatory conditions. They supress both humoral and cell-mediated responses. What must a doctor consider before placing a patient on a course of this medication? What are some possible consequences of long-term use?

Interaction of B and T cells in the adaptive immune system

As you learned in the previous section, when a macrophage encounters a foreign particle with an antigen attached to its surface, it surrounds and engulfs it, in the process of phagocytosis (Fig. 12.15). In the course of destroying the foreign material, the antigen that was present on its surface is moved to the surface of the macrophage, which then transports it to the lymph nodes.

The antigen-presenting macrophage is then presented to the helper T cell that has the T cell receptor corresponding to that particular antigen. This has the effect of activating the helper T cell.

The helper T cell can also be activated by B cells. When a B cell encounters the antigen that corresponds to its surface antibodies, it binds its antigens to its antibodies. It then processes the antigen, attaches it to its surface molecules and presents this to the helper T cells that have the matching T cell receptors.

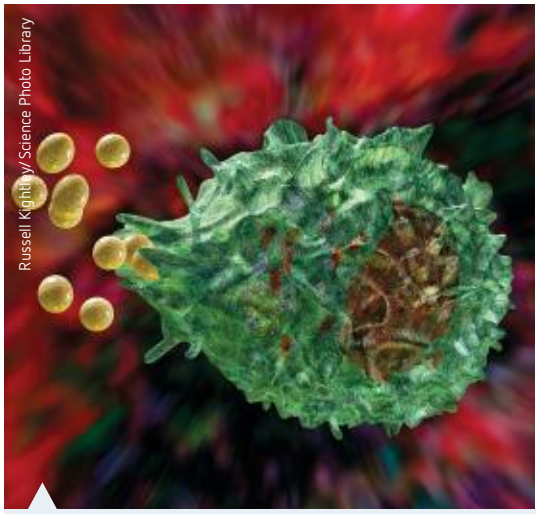

FIGURE 12.15 A macrophage (green) engulfing bacteria (yellow) by the process of phagocytosis

Worksheet
Chemical mediators and receptors of the immune response

Chemical signals in the form of cytokines are then secreted by the helper T cell to activate more of the same helper T cells and also macrophages. A specific cytokine chemical (interleukin-2) produced by the helper T cell activates the production of clones of the B cells that are specific to that antigen.

Interleukin-2 also activates the production of clones of the cytotoxic T cells that have that particular antigen receptor on their surface (Fig. 12.16).

When the immune response has successfully defeated the infection, suppressor T cells are responsible for suppressing the activity of the B cells and cytotoxic T cells.

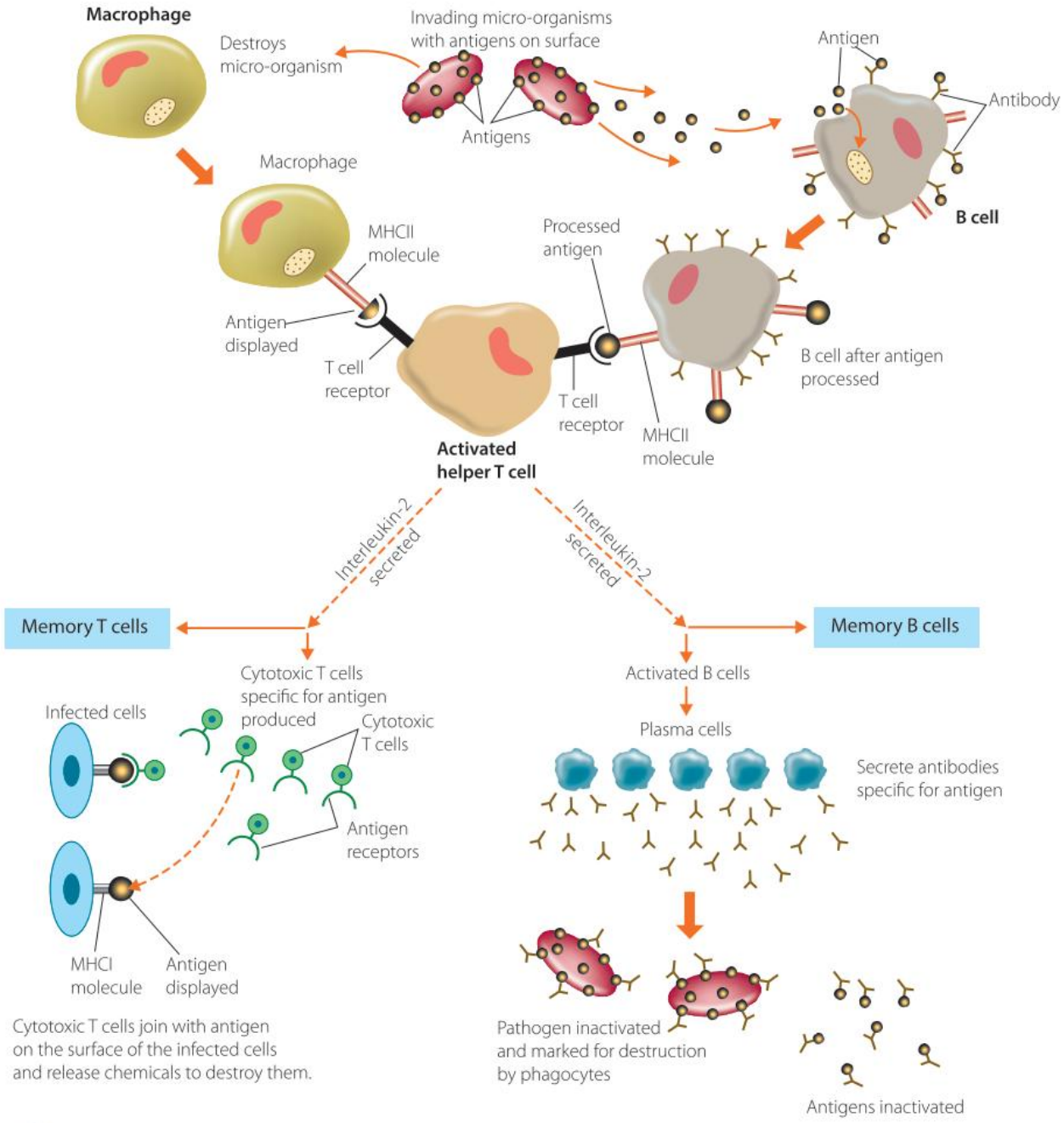

FIGURE 12.16 The immune response, showing interactions between B cells and T cells

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INVESTIGATION 12.1

A secondary source investigation into chemical signalling between the innate and adaptive immune systems

Information and communication technology capability

Literacy

In the past 10 years, there has been great progress in understanding the ways in which the immune system detects and responds to the presence of pathogens, as well as the ways in which the innate and adaptive immune systems communicate with each other. Communication between these two parts of the immune system is vital to an effective and coordinated response.

The mediators of the immune response generally act by the binding of molecules to surface receptors on host cells and pathogens. Others may have direct enzymatic effect or be directly toxic. The binding of a molecule to a receptor is a **stimulus** that elicits a **response** from a cell.

AIMS

1. To gather and process information on the ways in which cells of the innate and adaptive immune systems communicate with each other
2. To evaluate the importance of understanding chemical signals in the management of infectious disease

METHOD

1. Use a number of sources to research the following chemical mediators/receptors of the immune response:
   - **a** cytokines (e.g. interleukins, chemokines)
   - **b** complement proteins
   - **c** histamine
   - **d** prostaglandins
   - **e** leukotrienes
   - **f** PAMPs (pathogen-associated molecular patterns)
   - **g** TLRs (toll-like receptors)
   - **h** PRRs (pattern recognition receptors).

   When using a search engine, insert key words such as 'chemical mediators of inflammation' or simply the names of the molecules above.
2. Create a table to summarise your findings, including the following features:
   - **a** name of the chemical mediator or receptor
   - **b** its chemical nature (e.g. protein, lipid)
   - **c** its role in the response to pathogens.
3. Construct simple diagrams (using Word or other programs) to show how each of these features fits into the overall response to pathogen.
4. Prepare a references list to cite all your sources (page 27).

DISCUSSION

Evaluate the importance of understanding the chemical responses to pathogens. Of what benefit might this be to a scientist wishing to understand how to better manage an infectious disease? Justify this using an example of an infectious disease.

CONCLUSION

Write a summary sentence to evaluate the overall usefulness of understanding the chemical responses to infection.

INVESTIGATION 12.2

A practical investigation using the light microscope to examine the cells and tissues of the innate and adaptive immune systems

Ethical understanding

Personal and social capability

Work and enterprise

AIMS

1. To design and perform an investigation in order to obtain primary data on the structure and sizes of cells and tissues involved in the innate and adaptive immune systems
2. To assess and manage the risks involved in the use of appropriate technologies and biological materials

EQUIPMENT

- light microscope
- prepared slides of human blood smears, or images of these
- slides of human skin, sweat glands, lymph nodes, respiratory or intestinal epithelium, mucosa of the stomach, or images of these
- if available, prepared slides of pathogen-infected human tissues or images of these
- pencil and A4 paper
- phone camera to take photos of slides under microscope
- if available, digital microscope to project images of blood smears

Refer to Physics in Focus Year 11, Chapter 2 to revise how to use a light microscope.

RISK ASSESSMENT

- Use commercially prepared microscope slides of blood, to avoid the risk of biological contamination.
- Prepare a risk assessment table outlining safety precautions when using a light microscope.

METHOD

1. Review the correct technique for focusing an image under the light microscope. Evaluate your own technique and that of your peers as you proceed through this activity.
2. Place the tissue sample slide on the stage of the microscope and focus on low power.
3. Rotate to high power. Some sample images are provided on the next page, to help guide you in identifying the structures.
   - **a** goblet cells in respiratory and intestinal epithelium (Fig. 12.17)
   - **b** stratified squamous epithelium of the skin (Fig. 12.18)
   - **c** sweat and sebaceous glands in the skin (Fig. 12.19)
   - **d** gastric glands (acid producers) in the epithelium of the stomach (Fig. 12.20).
4. Use the images in figures 12.18 to 12.20 to help you identify a selection of structures that play a role in the innate immune system.
5. Record your observations in a table like the one in the Results section.
6. Place the blood smear slide on the microscope stage and focus on low power.
7. Use the images in figures 12.21 to 12.23 to help you identify a selection of structures that play a role in the innate and adaptive immune systems.

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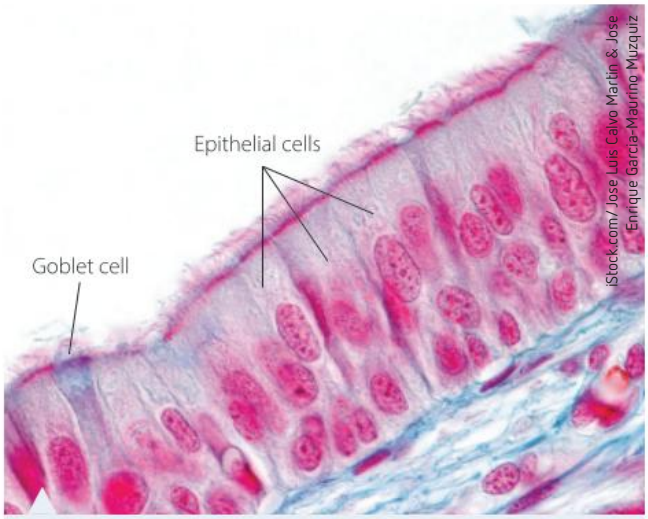


FIGURE 12.17 Goblet cells secrete mucus to trap pathogens that enter airways.

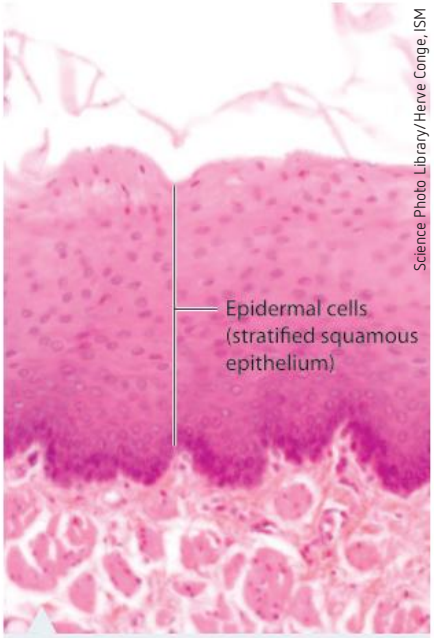


FIGURE 12.18 The epidermis consists of a waterproof external keratin layer as well as many layers of cells. This acts as a physical barrier to pathogens.

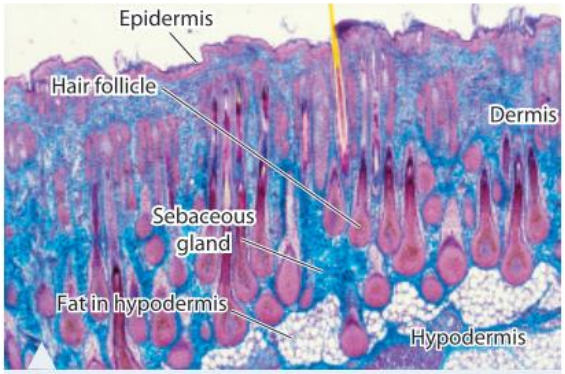


FIGURE 12.19 Oil and sweat both contain antimicrobial agents that form a chemical barrier to pathogens.

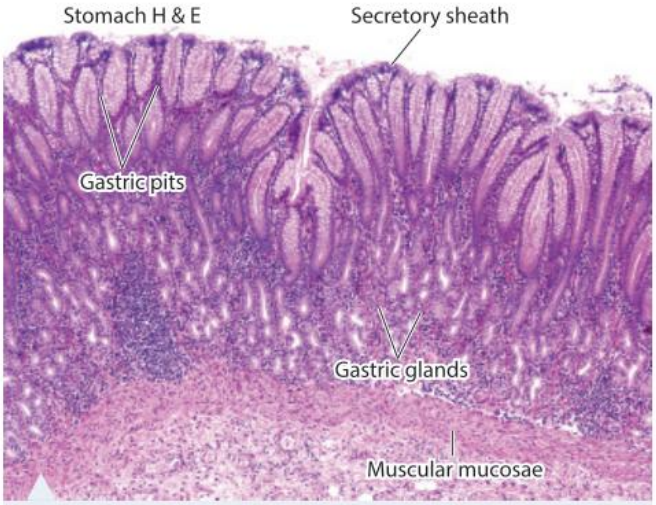


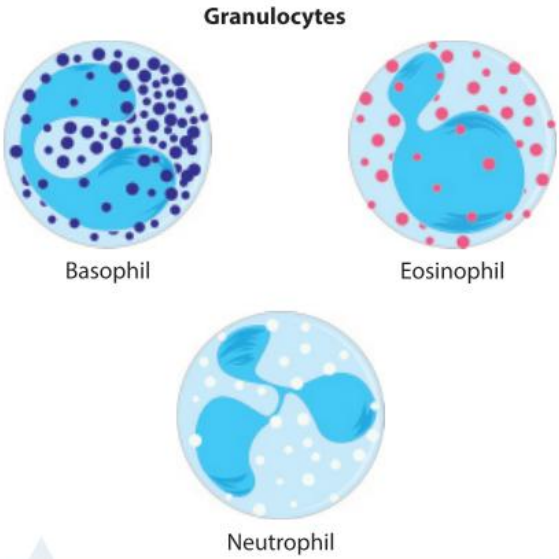
FIGURE 12.20 Stomach acid is produced by parietal cells. Acid is a chemical barrier to pathogens.

» 8 Record your observations in your table.

Some sample images of the structures are provided in Figures 12.21 to 12.23. Use these to help guide you in identifying the different blood cells.

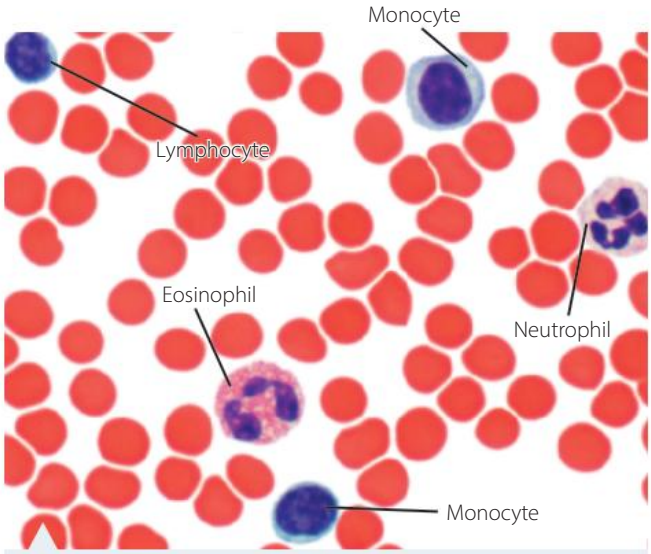
- **a** Innate immune system:
  - **i** neutrophils
  - **ii** eosinophils
  - **iii** basophils
  - **iv** monocytes/lymphocytes
- **b** Adaptive immune system:
  - **i** lymphocytes (B and T cells look the same under a light microscope)
  - **ii** plasma cells (Fig. 12.23).

9 Use secondary online and printed resources to research the role of each of these types of cells and tissues in the innate and adaptive immune response to pathogens.



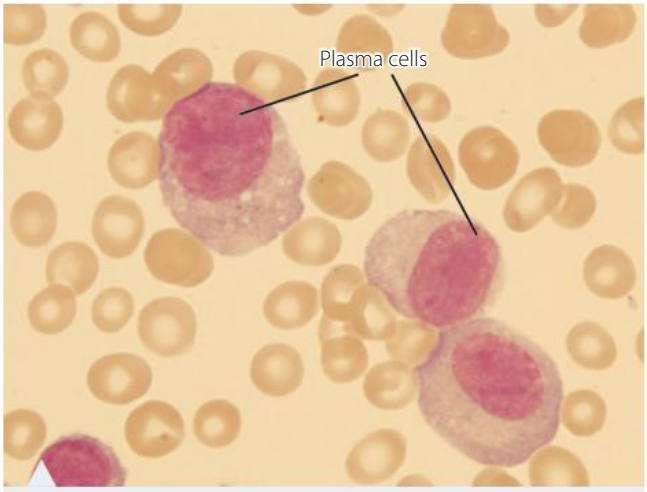
Microscape/Science Photo Library/iStock/toonishwarihead

FIGURE 12.21 Different types of white blood cells that are involved in the innate response to pathogens. Each is chemically attracted to sites of inflammation and plays a role in destruction of pathogens.



Steve Gschmeissner/Science Photo Library

FIGURE 12.22 Blood smear showing red blood cells and different types of white blood cells



LindseyRN/Shutterstock.com

FIGURE 12.23 Plasma cells are antibody-producing 'factories' and play a major role in the adaptive immune response.

RESULTS

Construct a table like the one below to record your observations.

CELL OR TISSUE TYPE	DESCRIPTION	DIAGRAM OR PHOTO	RESPONSE ON PRIMARY EXPOSURE TO A PATHOGEN

DISCUSSION

1. Describe some of the distinguishing features of the different cells involved in the innate and adaptive immune responses to pathogens.
2. Are cells and tissues actually coloured? Obtain some information from online and printed sources regarding two staining techniques that are used to help make these cells and tissues clearer – for example, Gram stain, H&E stain. Use a table to record their chemical nature and uses.
3. Imagine that you are a Biology teacher and you have been given the task of assembling a box of slides that represents a wide selection of cells and tissues involved in the innate and adaptive responses to pathogens. List some of the slides you would like to have in your collection. You may need to refer to other chapters in this book as a guide.
4. Which aspects of your microscope technique do you find most difficult? In what ways could you improve your approach to focusing a microscope?
5. What risks did you manage during this experiment?

CONCLUSION

Write a brief evaluation of the usefulness of the light microscope when examining tissue and cell samples involved in the innate and adaptive immune responses.

KEY CONCEPTS

- The two branches of the adaptive immune system interact via helper T cells.
- Both B cells and antigen-presenting macrophages can present antigens to helper T cells to initiate a response.
- Chemical signalling by helper T cells activates the cell-mediated and/or humoral response.
- Chemical signals are an important means of communication and coordination between the innate and adaptive immune systems.
- The light microscope is a useful technology for examining the cellular components of each system and understanding their distribution and effects in tissues.

CHECK YOUR UNDERSTANDING 12.1d

1. Justify the claim that the innate and adaptive immune responses do not act independently of each other.
2. Outline the role of helper T cells in adaptive immune responses.
3. Outline the purpose of memory cells (B and T) in adaptive immune responses.
4. How does the immune system ensure that cytotoxic T cells are produced that are specific for one type of pathogen?
5. What safety precautions are important when working with microscopes and prepared slides of biological material?
6. What features could you use to distinguish between neutrophils, eosinophils and basophils under the light microscope?

The immune response after primary exposure to bacteria

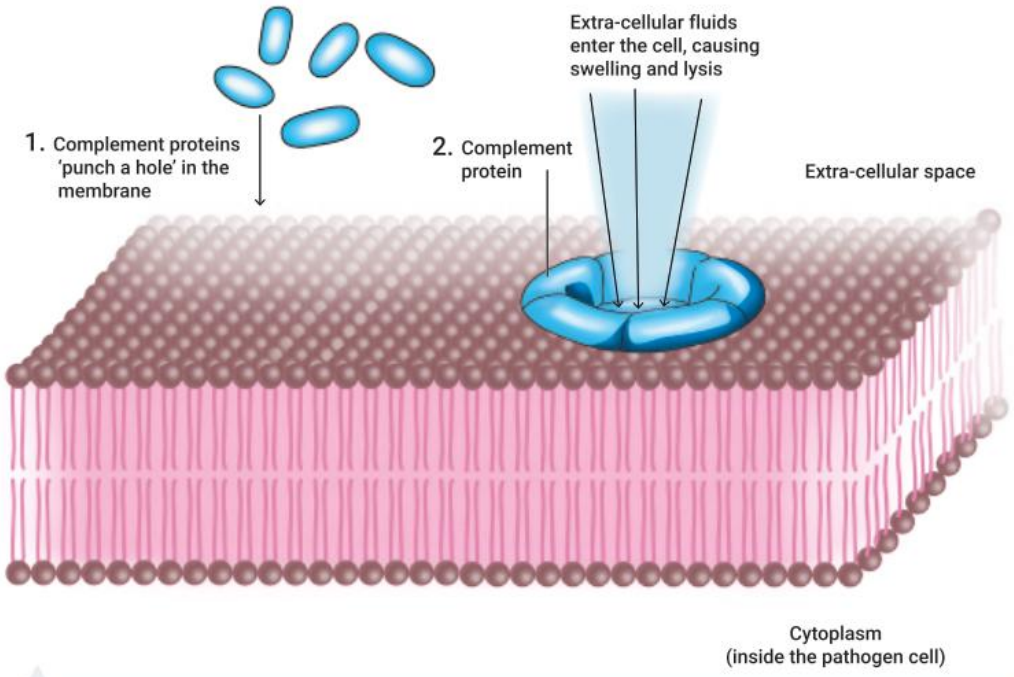
Bacteria are the micro-organisms that most frequently cause infections in humans. The primary response to bacteria includes natural barriers against infectious agents, as well as other innate responses and adaptive immunity.

The innate and adaptive responses to infection caused by bacteria are outlined below.

Innate immune responses

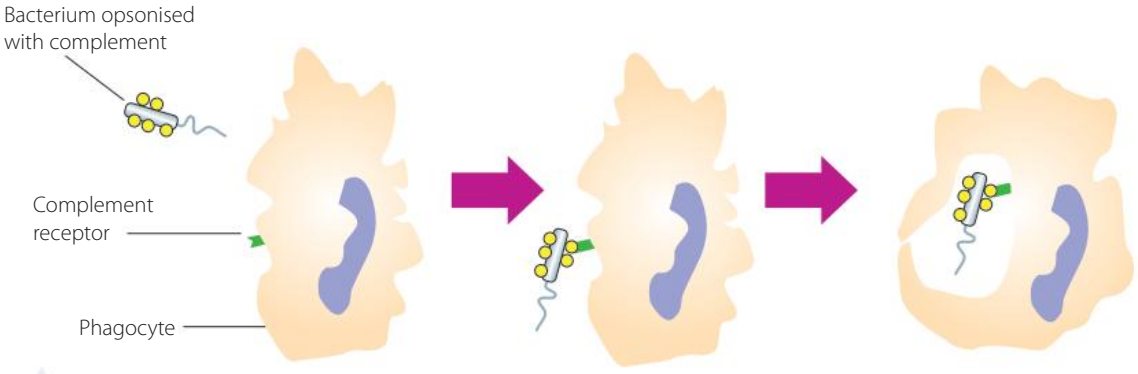
Innate immune responses to infection by bacteria include the following:

- Complement proteins directly puncture the bacterial cell wall and membrane and make it susceptible to osmotic lysis (Fig. 12.24).
- Opsonised bacteria (coated with antibodies and complement proteins) are phagocytosed (Fig. 12.25).
- Neutrophils are particularly active during bacterial infections. Peripheral blood levels of neutrophils increase in response to increased bone marrow production. This is especially common with



Wikipedia/Brazucs

FIGURE 12.24 Complement proteins puncture the bacterial cell membrane.



Source: Kerry Laing, Fred Hutchinson Cancer Research Centre, Seatle, USA. 'Immune Responses to Bacteria', Figure 2. British Society for Immunology.

FIGURE 12.25 Opsonised bacteria can be engulfed and destroyed by phagocytes.

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Gram-positive bacterial infections. Extreme infections may cause neutropaenia due to increased demand for neutrophils outstripping the ability of the bone marrow to supply them.

- Monocytosis (increased monocyte count) may occur, particularly in the resolution phase of a bacterial infection.

Adaptive immune responses

The adaptive immune response to infection by bacteria depends on whether the bacteria are intracellular or extracellular.

Intracellular bacteria

Cell-mediated immunity is launched against intracellular bacteria (such as *Salmonella*), which cannot be accessed by complement or antibodies. Infected macrophages present bacterial proteins on their cell surface using MHCII receptors. Helper T cells detect these and release interferon, which stimulates the macrophage to digest the bacterium-infected macrophage. This is an example of the innate and adaptive systems working together at the same time to solve the same problem (Fig. 12.26).

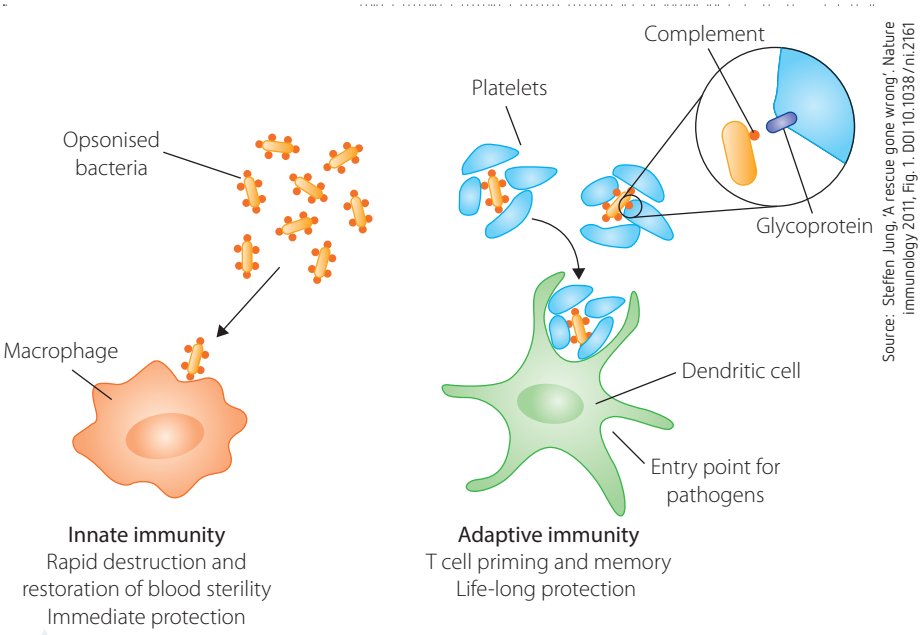


FIGURE 12.26 Opsonisation of bacterial cells for destruction by phagocytes involves both the innate and adaptive systems.

Extracellular bacteria

Extracellular bacterial infections (by *Staphylococcus aureus*, for example) are the most frequent of all. In such cases, the protection mechanisms are mainly related to the host's natural barriers, other innate immune responses and antibody production by the adaptive immune system.

TABLE 12.2 Responses of the immune system to bacterial infections

NATURAL BARRIERS	Structures/substances such as skin, mucous membranes, sebum, urine, gastric acid
INNATE IMMUNITY	• Complement proteins • Phagocytes (natural killer cells, neutrophils, macrophages)
ADAPTIVE IMMUNITY	• Antibody production by plasma cells • Cytokine production by T cells • Memory B cells formed

Case study: Immune responses to *Vibrio cholera* (cholera)

Every year, cholera affects millions of people in the developing world. The infection causes profuse diarrhoea and vomiting (Fig. 12.27). Death is caused by the resulting severe dehydration and shock due to fluid loss.

The bacterium that causes cholera is a curved or comma-shaped (vibrio) Gram-negative rod (Fig. 12.28).

The human immune system responds to primary exposure to *Vibrio cholera* in the following ways:

- innate response – production of cytokines such as interleukin, migration of neutrophils into the gut lining, and the production of antibacterial peptides (e.g. defensins) that kill bacteria directly or possibly alter host gene expression to induce more cytokines to induce inflammation
- adaptive response – IgA is secreted by the gut lining to protect it from bacterial colonisation. After a week there is an increase in circulating B lymphocytes specific to *V. cholera* antigens. Serum antibody peaks at 1–3 weeks after initial infection and decreases after about a year (Fig. 12.29). B memory cells form and provide a secondary response upon rechallenge at a later date with the same bacterium.

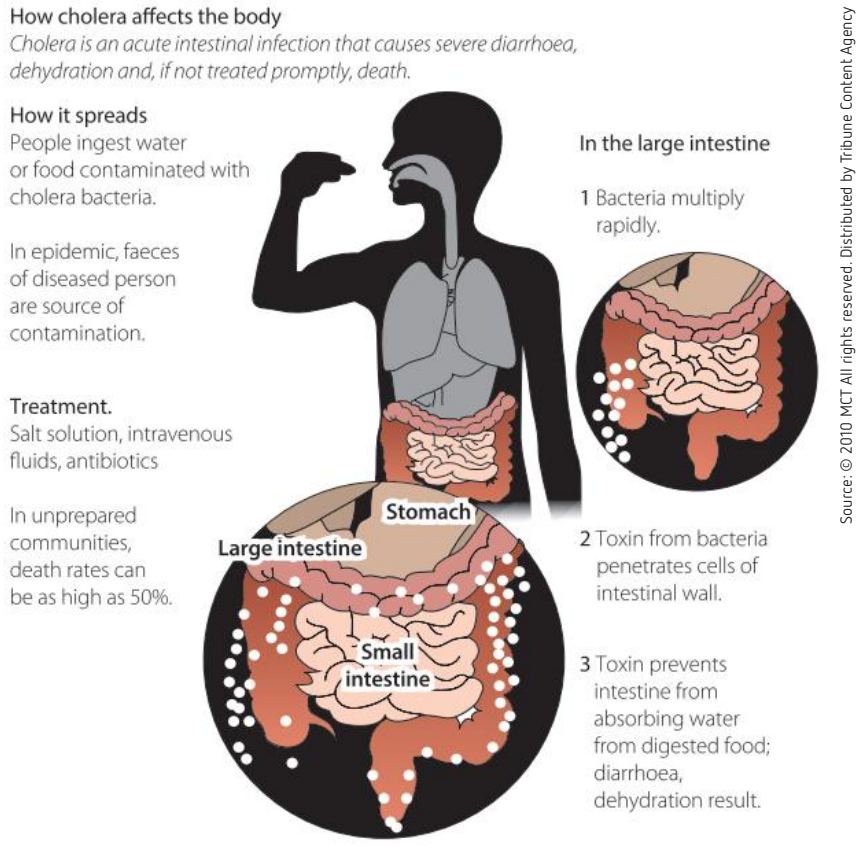


FIGURE 12.27 The pathogenic effects of cholera bacteria on the human gut

The microbiome of the gut plays a major role in regulating the immune response to diseases. All humans provide diverse microhabitats for an array of micro-organisms. After birth, all mammals begin to be colonised by foreign microbes. Symbiotic bacteria in the human gut have been found to prevent inflammatory disease during colonisation. There are probably extensive molecular interactions between the host and the bacteria that colonise the lower gut of humans.

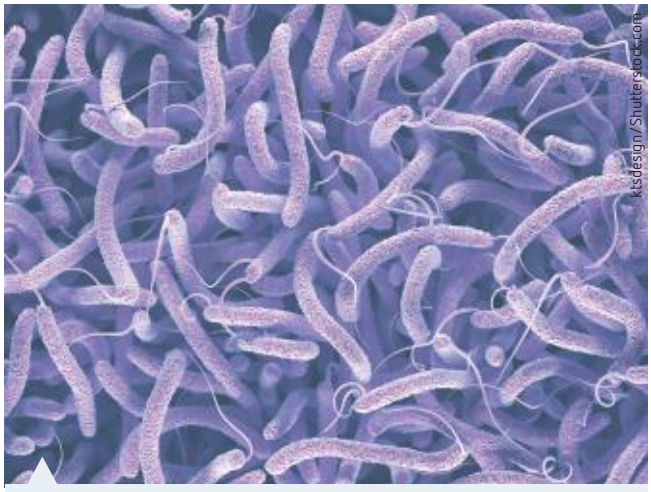


FIGURE 12.28 *Vibrio cholera* bacteria

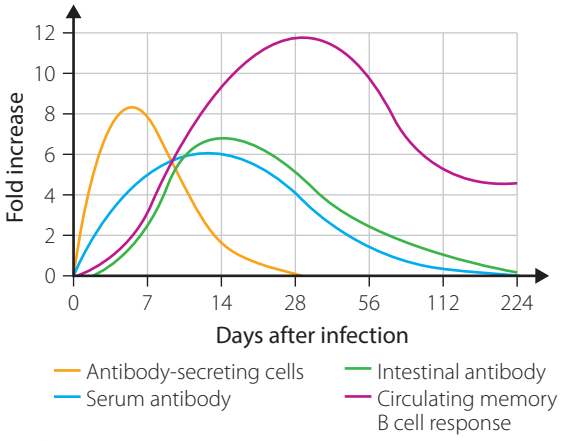


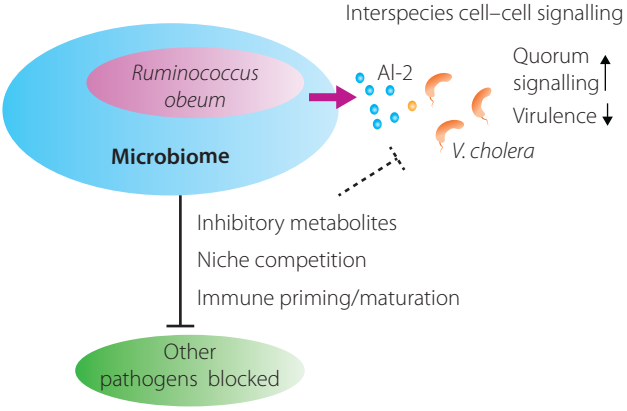
FIGURE 12.29 Timing and magnitude of adaptive responses after exposure to *Vibrio cholera*

One role of the microbiota is in providing **colonisation resistance** against pathogens. It does this by:

- competing for space and nutrients
- quorum sensing – this is the bacterial cell-to-cell communication that can occur between the pathogen and the microbiota (Fig. 12.30). For example, *R. obeum,* a gut microbe in humans, is able to sense interspecies signals and can block colonisation by producing inhibitory molecules (against, for example, *V. cholera*).

Understanding the mechanisms by which bacteria communicate with each other may be applied to restoring the health of people suffering from bacterial infections.

Progress has been made in understanding how the cholera bacterium may evade the body's defences. It has been discovered that the bacterium defends itself by attaching small amino acids to the large molecules, known as **endotoxins**, on its outer surface. These tiny amino acids change the electrical charge on the bacterial membrane from negative to neutral. The molecules that are produced to fight bacteria (CAMPs) are positively charged and generally bind to the negative bacterial surface, make a pore in the membrane and kill them. However, a neutrally charged bacterial membrane provides no opportunity for CAMPs to bind. *Vibrio cholera* are then able to invade the gut wall and cause the dramatic symptoms seen in cholera. Although an adaptive response follows, it does not generally reach a protective level quickly enough to enable the person to recover before the severe diarrhoea and vomiting kill them. The body's microbiota are also outcompeted in the process.



Source: Amanda J. Hay, Jun Zhu, 'Microbiota Talks Cholera out of the Gut', Fig. 1 Cell Host & Microbe Vol. 16, Iss 5, November 2014. Elsevier

FIGURE 12.30 Complex chemical signalling occurs between the microbiome and pathogens. AI-2 is an inhibitory molecule produced by the microbiota in response to infection.

If a drug could be developed that disables these amino acids, CAMPs could do their work and reduce the invasiveness of the bacteria. The innate immune system would prevail.

The immune response after primary exposure to fungi

Humans are constantly exposed to a wide range of fungi through digestion, inhalation or traumatic injury. Most fungi cause no disease in humans. Superficial fungal infections of the skin (dermatophytes) are common.

Innate immune responses

The innate immune system is heavily involved in the response to fungal pathogens. Physical barriers such as the skin and mucous membranes provide a first-line barrier. Phagocytes such as dendritic cells, neutrophils and macrophages are the main cells that use pattern recognition receptors (PRRs) on their surface to recognise and bind to fungi. An increase in neutrophils in the peripheral blood is common.

Adaptive immune responses

Adaptive immune responses to fungi are poorly understood. Cell-mediated and humoral immunity play a role.

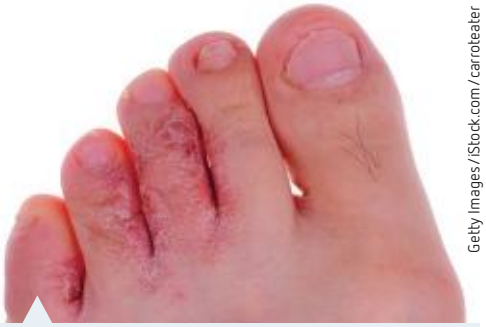
- Phagocytosis is initiated and alerts effector cells to respond via cytokine production-induced inflammatory responses.
- Dendritic cells transport ingested fungi to the lymph nodes, where helper T cells and memory T cells are recruited.
- Helper T cells produce interleukin-17 and further stimulate neutrophils and macrophages. Interleukin-17 is critical in antifungal immunity – it promotes inflammation.

Case study: Immune responses to *Trichophyton rubrum* (tinea)

Dermatophyte infections are common around the world. They are the main form of fungal infections of the body skin (tinea corporis), feet (tinea pedis or 'athlete's foot') and nails (tinea unguium).

Tinea pedis causes scaling, redness and itching of the feet and toes (Fig. 12.31). It is most common in South-East Asia, Africa and parts of Australia, probably due to lack of enclosed footwear.

There is evidence that *T. rubrum* is able in some cases to evade the initial innate immune system responses, and therefore the adaptive T cell responses are never activated. Both are necessary to clear the infection.



Getty Images/iStock.com/carroteater

FIGURE 12.31 Tinea pedis, caused by the fungus *Trichophyton rubrum*

In host tissues infected by *T. rubrum*:

- enzymes called keratinases, which break down the skin barrier, are secreted
- molecules in the fungal cell wall, called mannans, inhibit the immune response and reduce replacement of superficial skin cells
- a hypersensitivity response (immediate or delayed) is elicited.

Responses of the immune system:

- Keratinocytes express TLRs (toll-like receptors) and defensins as a first-line response.
- Dendritic cells are primarily responsible for regulating the response to tinea – they suppress fungal growth and stimulate cytotoxic T cell production. Therefore the response to tinea is mediated by the innate and adaptive (cell-mediated immunity) responses.
- PRRs recognise PAMPs, and so phagocytes express a mannose receptor that recognises mannans in the fungal cell wall. Cytokines are released, and phagocytosis of fungal cells occurs, with proliferation of cytotoxic T cells.

The infection becomes chronic in 20% of patients (probably due to a defective cell-mediated response). In patients with chronic infections, depressed cytotoxic T cell responses are seen. It is suspected that

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some fungi are able to evade the mannose receptors on the cell membranes of epithelial cells and avoid recognition, which means no cytokine is released and no phagocytosis is induced, and T cells are inhibited. An increase in neutrophils (neutrophilia) and monocytes (monocytosis) may be seen in a full blood count (a blood test that shows the numbers of different types of blood cells). This is an example of the importance of the interaction between the innate and adaptive immune systems. Without the initiation of the innate responses, the adaptive responses are not activated and infection continues unabated.

The immune response after primary exposure to viruses

Viruses must invade the cells of a host in order to replicate. Once the virus is inside a cell, the host's immune system has no access to the virus itself. Therefore, the infected cells use MHCI molecules on their cell membrane to present pieces of viral protein to the outside of the cell.

As part of the adaptive immune system, cytotoxic T cells recognise specific virally infected cells. Only T cells with the specific T cell receptors for that particular virus are activated. The cytotoxic T cell then releases cytotoxic factors that kill the virally infected cell.

Some viruses are capable of stopping the MHCI molecules from migrating to the cell surface and presenting viral protein. If this occurs, the immune system doesn't know the cell is infected. However, the innate immune system has found a way around this. Natural killer cells (phagocytes) will detect cells with fewer than normal MHCI receptors and release cytotoxic factors, killing them as they would a virally infected cell. These cytotoxic factors include **perforin**, which makes pores in the cell membrane, and **granzymes**, which enter through the pores and induce programmed cell death (apoptosis) in the virally infected cell. Cytotoxic T cells also produce cytokines (such as interferon) that prevent the replication of viruses inside the infected cells. In addition, interferon signals neighbouring cells to increase the number of MHCI molecules on their surface, to act as a signal for T cells.

Humoral immunity can play a role in responding to viruses before they infect cells. Antibodies are produced by B lymphocytes. The virus becomes coated in antibodies and is no longer capable of infecting cells. It is also a target for phagocytes. Antibodies also activate the complement system, and the complement proteins bind to the viral envelope and damage it.

Full blood counts can be unpredictable in viral infections. The white cell count may be reduced (leukopaenia) due to the suppression of bone marrow production of white cells by the virus. Sometimes this is due to an increase in lymphocytes and monocytes but a greater decrease in neutrophils.

Case study: Immune responses to the Dengue fever virus

Dengue fever is a worldwide threat that affects millions of people every year. The virus that causes Dengue is transmitted by the mosquito *Aedes aegypti*. It is most common in tropical countries.

Symptoms of the disease include fever, headaches, rash, vomiting, and joint and muscle pain. Pain behind the eyes (retro-orbital pain) is a common feature. The disease commonly resolves after 1–2 weeks. In fatal cases, death usually occurs due to haemorrhage because of a severe lack of platelets.

The Dengue fever virus infects cells of the immune system itself. This is part of its insidious nature. The innate immune system is targeted, as the virus infects macrophages and dendritic cells. Once the innate immune system is evaded, the virus becomes blood-borne and spreads around the body. It can then be transmitted to another host when a mosquito takes a blood meal from the infected patient.

The usual situation with viral infections is that dendritic cells recognise viral PAMPs and release interferons. Interferons activate the adaptive immune system, which leads to both T and B cell responses.

In some people, upon infection with the Dengue virus, certain types of interferons (type I IFN) are not produced by the dendritic cells. This means they are unable to signal helper T cells to become activated. There is also evidence that any type I IFN that is released has its signalling mechanisms blocked by the virus. This is yet another example of the intricate signalling and cooperation that occurs between the

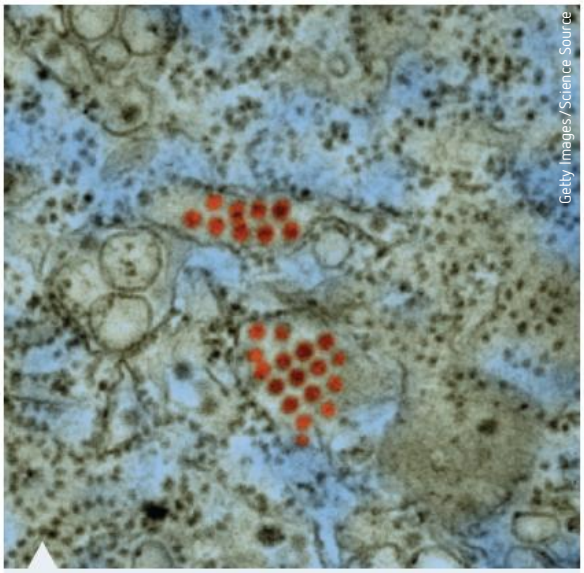


FIGURE 12.32 Dengue fever virions inside host tissue

innate and adaptive responses. It explains the clinical observation that the innate and adaptive immune responses are very poor in Dengue patients.

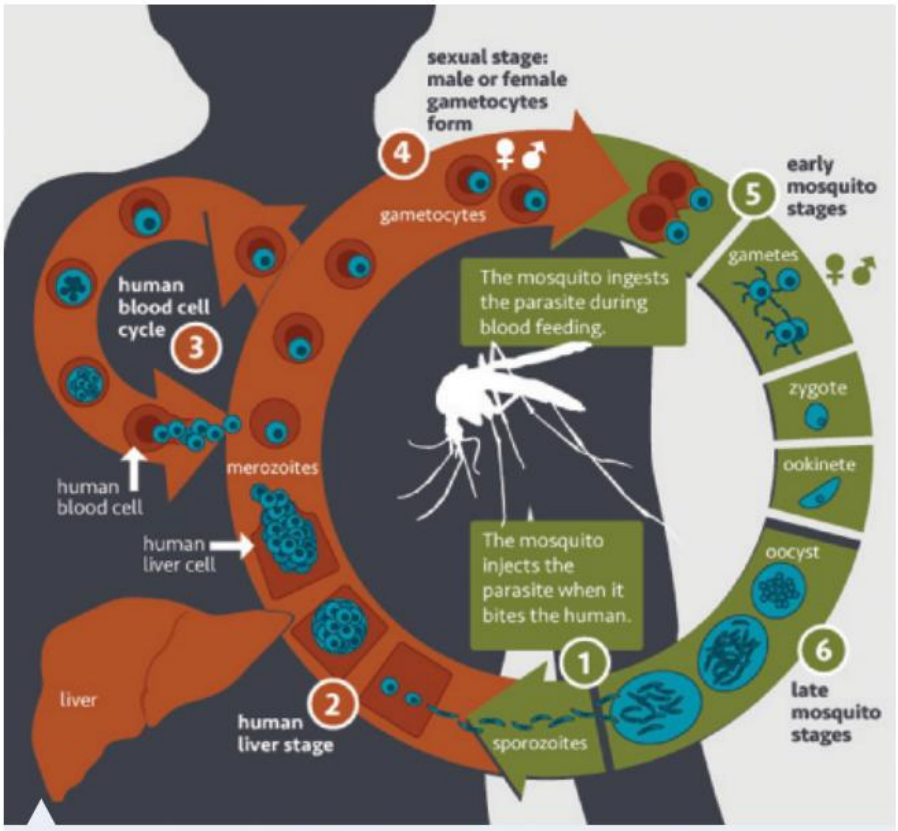
In addition, Dengue fever virus is able to form intracellular membrane pockets/vesicles to hide in and replicate before any PAMPs are detected by the host cell's PRRs.

During a primary infection, antibodies are raised against the particular virus. There is some evidence that these antibodies are cross-reactive against other **serotypes** of the Dengue virus. This only occurs when the antibody concentration is high, as cross-protection wanes quickly. People may have type-specific antibodies from a Dengue fever infection for decades. B memory cells as well as long-lived plasma cells are generated by an infection with this virus.

In some patients, the T cells react very quickly and may actually enhance the severity of the symptoms. This is of concern in the development of a vaccine.

The immune response after primary exposure to protozoa

Protozoa are a group of protists that display complex multi-stage life cycles that make launching an immune response problematic. Figure 12.33 shows the life cycle of malarial protozoa.



National Institutes of Health, National Institute of Allergy and Infectious Diseases. Creative Commons Attribution 2.0 Generic (CC BY 2.0). https://creativecommons.org/licenses/by/2.0/.

FIGURE 12.33 Life cycle of the malaria parasite, *Plasmodium*

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Protozoa usually cause chronic infections, because innate immune defences are often ineffective and the pathogen has evolved many mechanisms to resist adaptive immune defences.

Protozoa activate different immune responses than those activated by bacteria, viruses and fungi.

- Full blood counts usually show eosinophilia (increased number of eosinophils), because of accelerated bone marrow production of eosinophils.
- Although protozoa may be phagocytosed by macrophages, many are resistant to phagocytic killing and may even replicate within macrophages.

Protozoa also possess features known as *escape/evasion mechanisms*, which are strategies by which parasites avoid the killing effect of the immune system in an immunocompetent host. Escape mechanisms used by protozoal parasites include the following ways of 'fooling' the immune system:

- *antigenic masking* – escaping immune detection by covering itself with host antigens
- *blocking of serum factors* – acquiring a coating of antigen–antibody complexes or non-cytotoxic antibodies that sterically (by arrangement of the atoms) blocks the binding of specific antibody or lymphocytes to the parasite's surface antigens
- *intracellular location* – the intracellular habitat of some protozoan parasites protects them from the direct effects of the host's immune response; by concealing the parasite antigens, this strategy also delays detection by the immune system
- *antigenic variation* – changing its surface antigens during the course of an infection; parasites carrying the new antigens escape the immune response to the original antigens
- *immunosuppression of the host* – the maturation of dendritic cells and monocytes is inhibited in red blood cells infected by *Plasmodium falciparum*. This reduced immune response may delay detection of antigenic variants. It may also reduce the ability of the immune system to inhibit the growth of and/or kill the parasites.

Case study: Immune responses to *Plasmodium* spp. in malaria infections

People who are exposed to the malaria parasite *Plasmodium falciparum* develop naturally acquired immunity. The details of how this works are still unclear. Many people in endemic areas carry a large population of parasites in their blood but experience almost no ill effects. These levels of infection would be fatal to a newly exposed person. Another complication is the fact that many of the surface proteins of the protozoa show great antigenic diversity. A host would need a vast repertoire of antibodies to cope with every variation available.

The immune responses to infection with malarial parasites are outlined below.

Innate immune responses

- Some individuals are inherently protected from the parasite due to a change in the structure of their haemoglobin or red blood cells – examples are people with sickle cell anaemia, some thalassaemias, or Duffy negative red blood cells.
- Natural killer (NK) cells increase in number; they cause lysis of parasite-infected red blood cells. NK cells produce interferon, leading to macrophage activation. NK cells also induce the production of chemokines, which activate phagocytes and help to limit the liver stage of the parasite.

Adaptive immune responses

- Antibodies (especially IgM and IgG) are produced by plasma cells.
- Helper T cells and cytotoxic T cells play a role in parasite control.
- Repeated infections lead to different stages of immunity, ranging from anti-parasite immunity (prevents the parasite getting into the blood) to anti-disease immunity (no clinical signs observed despite persistent infection), to **premonition** (protection from new infections due to a low-grade infection of the blood with malaria parasites).

- A merozoite is a stage of the *Plasmodium* life cycle that multiplies inside red blood cells. Antibodies may inhibit merozoite invasion of erythrocytes or enhance the elimination of infected erythrocytes in the spleen.
- Opsonisation leads to increased susceptibility to phagocytosis, destruction by cytotoxic T cells or parasite inhibition by neutrophils and macrophages.
- Acquired immunity does not last long, and if a person leaves the area they become vulnerable once more.

The immune response after primary exposure to macroparasites

Parasitic worms (**helminths**) are a diverse group of antigenically complex organisms that infest millions of people worldwide every year. Infestation by parasitic worms is called helminthiasis (Fig. 12.34). Most people who are infested with worms experience few signs that they are infested, but some people suffer from life-threatening consequences. It is mostly a problem of developing countries.

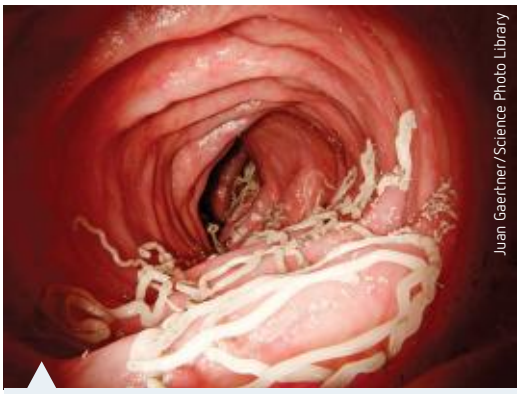

FIGURE 12.34 A segment of intestine infested with intestinal worms

Pathogen factors

Pathogen factors pose unique challenges for the adaptive and innate immune responses to macroparasites.

- Many parasitic worms have multiple life stages.
- Many worms infest a number of hosts as they develop. They change dramatically before being transmitted to the next host.
- Chronic infestations can lead to pathological changes.
- Parasites may modulate their surface structure to avoid recognition by the host.
- Molecular cross-talk occurs between helminths and the mammalian immune system (that is, the parasite uses similar immune signalling molecules to those of the host); schistosomiasis is an example.

Host factors

Immune responses are predictable, with increases in interleukin, IgE, eosinophils and mast cells detectable upon infestation.

- Full blood counts usually show eosinophilia in response to accelerated bone marrow production of eosinophils.
- Th2 cytokines such as interleukins are increased.
- Immune response modulation – the initial increase in T cell reaction dampens as the infestation becomes chronic. This is in the best interests of the host.
- Memory protects the host from new infection while the old infestation continues. One possibility is that the adult parasites evade the immune system. Alternatively, the primary infestation may alter the anatomy/physiology of the host to prevent further infestations.
- Antiparasite IgE levels correlate with resistance to new infestations in the host.
- Th2 response genes in some families help confer resistance – this is closely linked with the ability to expel the parasite.
- Eosinophils kill opsonised helminths.

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INVESTIGATION 12.3

Changes in the blood during infectious disease

Information and communication technology capability

Literacy

As part of a diagnostic routine, a doctor may ask for a full (or complete) blood count to be taken from a patient with a suspected infectious disease. The blood count will determine absolute numbers and ratios of white and red blood cells as well as their features (sizes, shapes, **inclusion bodies** in the nucleus or cytoplasm). Although this does not always give a definitive diagnosis, and certainly does not identify the specific pathogen, it can alert the doctor as to how the immune system is responding. Different types of pathogens generally create different patterns of white and red blood cell changes. The test can also indicate how long the infection has been present, whether the infection is overwhelming the body's defences, and whether the immune system is mounting an appropriate response to the infection.

The number of circulating white cells in the blood reflects a balance between their use (in responding to infections) and the rate of their production by the bone marrow. Any process that changes one of these will change the dynamic equilibrium between them and result in an increase or decrease in white cells (Fig. 12.35).

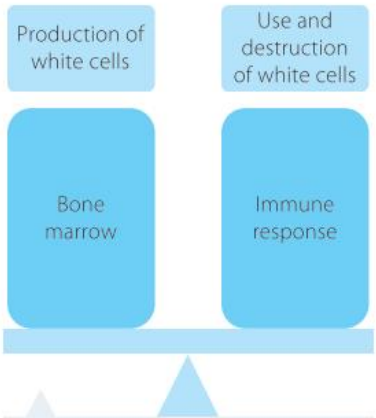

FIGURE 12.35 The blood level of white cells reflects both production and use levels.

When the total count of a white cell increases, generally the suffix 'philia' is added to the name of the white cell. For example, an increase in neutrophils is referred to as 'neutrophilia'. For some cells, the suffix 'cytosis' means increased production. For example, monocytosis refers to increased production of monocytes. When white cells decrease, the suffix 'paenia' is added to the name. For example, neutropaenia indicates a lower than normal number of neutrophils in the peripheral blood.

A typical pathology report based on a full blood count looks something like the one in Figure 12.36.

COMPONENT	YOUR VALUE	STANDARD RANGE	UNITS	FLAG
White blood cell count	5.4	4.0 – 11.0	K/uL	
Red blood cell count	5.20	4.40 – 6.00	M/uL	
Haemoglobin	16.0	13.5 – 18.0	g/dL	
Haematocrit	47.2	40.0 – 52.0	%	
MCV	91	80 – 100	fL	
MCH	30.8	27.0 – 33.0	pg	
MCHC	33.9	31.0 – 36.0	g/dL	
RDW	12.7	< 16.4	%	
Platelet count	*149*	*150 – 400*	*K/uL*	*Low*
Differential type	Auto			
Neutrophil %	56	49.0 – 74.0	%	
Lymphocyte %	*23*	*26.0 – 46.0*	%	*Low*
Monocyte %	*13*	*2.0 – 12.0*	%	*High*
Eosinophil %	*7*	*0.0 – 5.0*	%	*High*
Abs. Neutrophil	1	0.0 – 2.0	%K/uL	
Abs. Lymphocyte	3.1	1.0 – 5.1	%K/uL	
Abs. Monocyte	1.2	0.0 – 0.8	%K/uL	
Abs. Eosinophil	0.7	0.0 – 0.5	%K/uL	
Abs. Basophil	0.0	0.0 – 0.2	%K/uL	

FIGURE 12.36 A typical full blood count with a differential count of white blood cells

AIMS

1. To collect valid and reliable secondary data on typical changes in the blood profile for different infectious diseases
2. To analyse and evaluate sec ondary data and information on full blood counts

HYPOTHESIS

Write a suitable hypothesis for your investigation.

METHOD

1. Use a number of sources to research the changes in white blood cells during bacterial, fungal, protozoal, viral and macro-organism infections.

   When using a search engine, insert key words such as 'white cell changes in bacterial infections'.
2. Using the information you have gathered, create a table to record the following data:
   - **a** the type of pathogen
   - **b** any changes in the total number of white cells
   - **c** any other unusual features of the white cells – for example, more immature forms (can indicate bone marrow response to infection through increased WBC production), strange shapes, structures inside nucleus or cytoplasm ('inclusion bodies')
   - **d** explanation for the changes in the particular cells based on their primary role in responding to pathogens
   - **e** any changes in red cells (e.g. numbers, shape or increased nucleated red cells)
   - **f** include the 'normal range' for these cells (usually given in brackets on RHS of report)
   - **g** Make a small table and record the scientific units in the table. Find out what the symbols stand for.
3. Include a reference list of websites or any other resources used.

DISCUSSION

1. Evaluate the usefulness of the full blood count as a diagnostic tool for doctors when they suspect a patient has a bacterial infection.
2. Sometimes there are no changes to the peripheral blood during an infection. How might this be explained?
3. Find out the clinical significance of the following:
   - **a** neutrophila with a 'left shift'
   - **b** toxic neutrophils
   - **c** neutrophils with Dohle bodies.
4. What types of pathogens generally cause eosinophilia?
5. What types of pathogens generally cause neutrophilia?
6. How common is basophilia?

CONCLUSION

Write summary sentences related to the aim of this investigation.

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KEY CONCEPTS

- The immune system responds to extracellular bacterial infections by producing increased numbers of neutrophils; complement proteins punch holes in bacterial cell walls.
- Intracellular bacteria are dealt with by cytotoxic T cells.
- The gut microbiome plays a key role in defence against pathogens such as the cholera bacterium.
- Fungal infections are mostly prevented by barriers such as the thick keratin layer of the skin.
- Phagocytes engulf invading fungi; cytotoxic T cells may become involved; neutrophilia and monocytosis may be seen in the full blood count.
- Leukopaenia (reduced white cell count in the blood) is a common response to viral infections.
- Protozoa typically cause eosinophilia in response to accelerated bone marrow production of the cells.
- Some protozoa have strategies to evade the immune response.
- Macroparasites such as worms cause eosinophilia and increased IgE levels in the peripheral blood.
- A full blood count is a useful start in identifying the presence of pathogens in the body but is not accurate when used on its own.

CHECK YOUR UNDERSTANDING 12.1e

Choose *one* of the pathogen case studies presented in the preceding section and answer the following questions for that pathogen.

1. Identify the scientific name of the pathogen.
2. Describe the effects of this pathogen on the host.
3. Outline the barriers that are present to prevent entry by this pathogen.
4. Describe the primary cellular responses of the innate immune system to the presence of this pathogen.
5. Describe the primary cellular responses of the adaptive immune system to this pathogen.
6. What chemical responses help to mediate the responses of cells to the presence of this pathogen?
7. Outline the types of changes in the full blood count you might expect to see when this pathogen has been present in the body for some time.

12.2 Modelling the innate and adaptive immune systems

In science, a model is a representation of an idea, an object or even a process or a system. The model is used to describe and explain phenomena that cannot be experienced directly. Models are central to what scientists do, in their research and when communicating their explanations.

Models are a simplified representation of an imagined reality. They are used to link theory with experiment, by guiding research and enabling predictions to be developed and tested experimentally.

Models have always been important in science, and continue to be used to test hypotheses and predict information. Often they are not accurate, because the scientists may not have all the data. It is important that scientists test their models and be willing to improve them as new data becomes known. It can take time to get a model right.

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FIGURE 12.37 A model of the DNA double helix shows important component parts and how they fit together.

Worksheet
Cells of the adaptive immune system

INVESTIGATION 12.4

A practical investigation to model the responses of the innate and adaptive immune systems

Information and communication technology capability

Literacy

Personal and social capability

You are to model one of the three main responses of the immune system to invading pathogens. This includes the cellular and chemical components of each system, as well as the different types of pathogens dealt with (e.g. intracellular versus extracellular; toxins versus viruses).

AIM

To model the components and responses of the human immune response, to explain the immune response to pathogens

METHOD

Weblink
Modelling the immune system

1. You will work individually or in a group, and become 'expert' in one of the types of responses of the human immune system.
2. Use online or printed resources available to you.
3. Choose *one* of the following three immune responses:
   - innate responses (first or second line) (you may choose one aspect, e.g. chemical barriers)
   - adaptive response – humoral
   - adaptive response – cell-mediated.
4. Choose a form for your model. Here are some suggestions:
   - a 3D model of the cells in both of the systems made from various materials (e.g. playdough)
   - a computer animation using the software of your choice (e.g. PowerPoint, Blender, Poser, Adobe After Effects, Go Animate, Minecraft)
   - a 2D model, such as a poster
   - a board game that incorporates the cells and their functions
   - a role play
   - a story board or series of cartoons
   - There are also a number of suggestions for immune system models and games on the Internet. Use the search words 'simulate immune system + student'. Also consider suggestions from other students or your teacher.

Weblink
Acting out the immune response

5. Your model should demonstrate an accurate understanding of the following:
   - Identify the specific pathogen (Chapter 10) or other antigen (e.g. toxin).
   - Identify the chemical and cellular components of the immune response.
   - Outline the circumstances that trigger the response.
   - Indicate the speed of the response (minutes, hours, days).
   - Indicate the presence or absence of memory formation from this response.
   - Give some indication of the site of these reactions (e.g. in tissues, in lymph nodes).

Weblink
A system of many hats

6. When completed, your model can be presented to the class.
7. All students should complete the summary below when all models have been presented.
8. Create a bibliography/reference list in the appropriate format (page 27).

»

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RESULTS

Copy and complete the following table individually after all models have been presented.

FOCUS AREAS	INNATE RESPONSE	ACQUIRED HUMORAL RESPONSE	ACQUIRED CELL-MEDIATED RESPONSE
What initiates the response?			
How fast is the response?			
Cellular components			
Chemical components			
Presence or absence of memory formation (+/–)			

DISCUSSION

1. In what ways did modelling make it easier to explain a concept to others?
2. What are some of the limitations of the model you created (what *couldn't* it explain)?
3. How important is the role of models in science education? Why are models of particular value to science?

EXTENSION

There are a number of excellent modelling activities available online. The weblink provides examples of the wide range of ways in which immune responses can be modelled by a class.

KEY CONCEPTS

- Models are used by science to explain concepts in simple and effective ways.
- Models often allow scientists to make predictions.
- Models may be in the form of physical structures or mathematical equations.
- Models are constantly changing as new information comes to light.

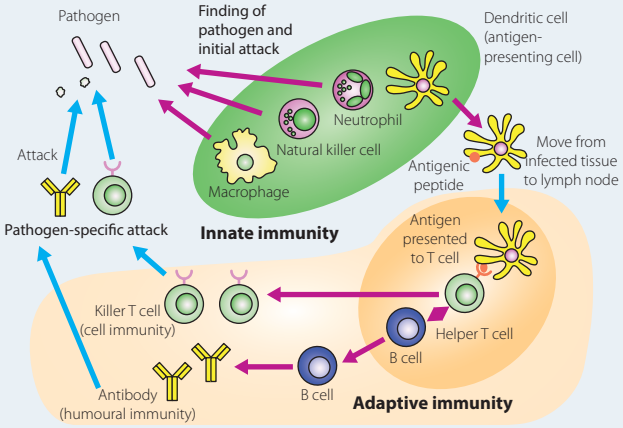
CHECK YOUR UNDERSTANDING 12.2

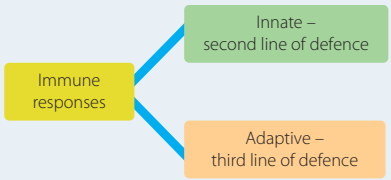
1. Outline some of the features of a good scientific model.
2. Suggest reasons why a physical model of a scientific concept may be more appropriate than a written explanation in certain circumstances.
3. What are some of the challenges faced when designing a model for something as complex as the innate and adaptive immune responses?
4. Describe some of the ways in which you evaluated your own model and made improvements during your investigation.
5. Discuss the value of peer assessment and review of scientific models. In what ways did this assist or hinder your model making?
6. Think of two established scientific models you have studied in any Science class. What features of these models have made them so successful and enduring?

12 CHAPTER SUMMARY

Immunity: How does the human immune system respond to exposure to a pathogen?

INNATE AND ADAPTIVE IMMUNE SYSTEMS WORK TOGETHER



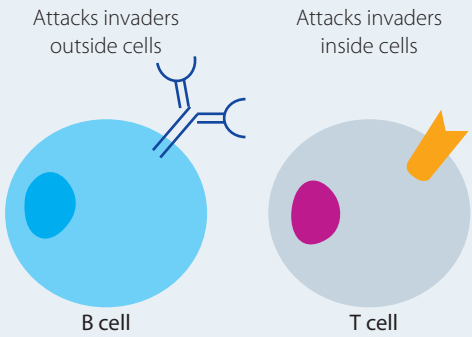


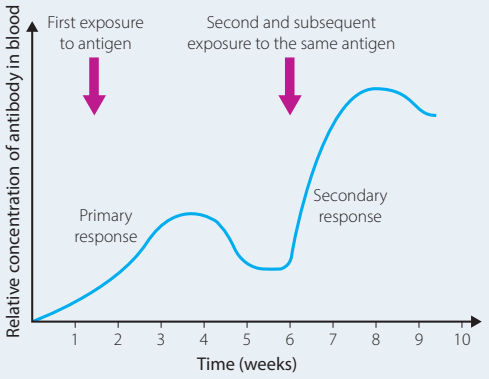
Third line of defence – adaptive immunity

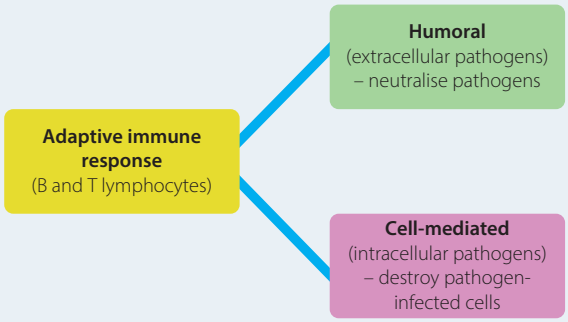
B and T lymphocytes are capable of recognising foreign molecules, called *antigens* ('antibody generators'). Antigens are components of bacteria, viruses, fungi, worms, tumour cells and other non-cellular materials.

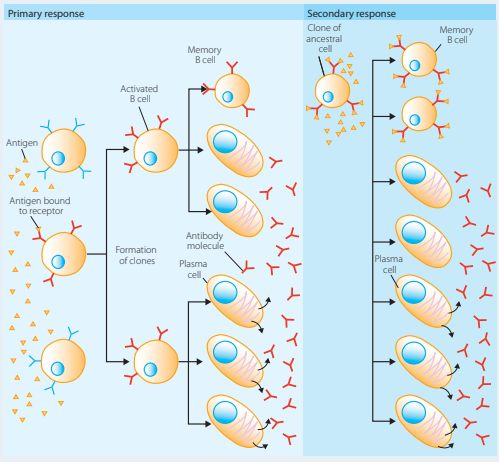
The adaptive immune response:

- is specific
- has a diverse range of responses
- has memory
- has self-tolerance.









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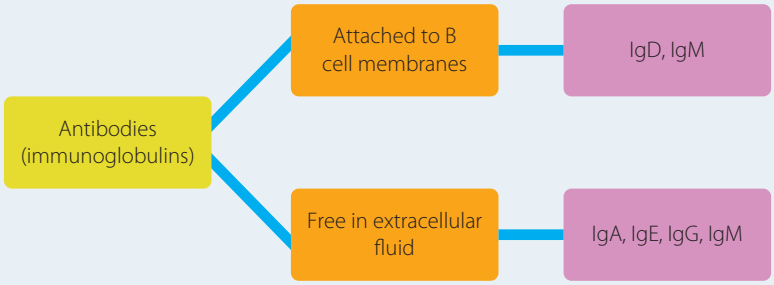
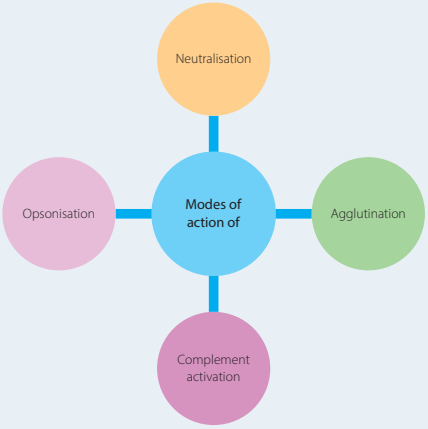
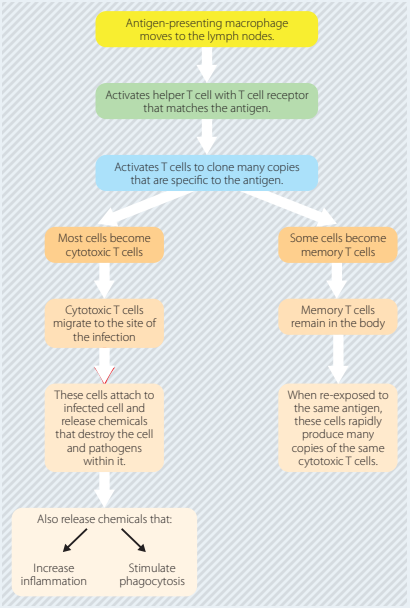


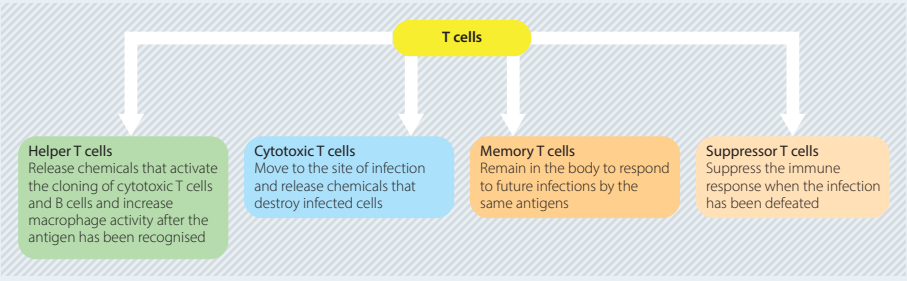


The human body recognises *patterns* of self and non-self – this is the basis of all immune responses.



Common responses to different pathogens

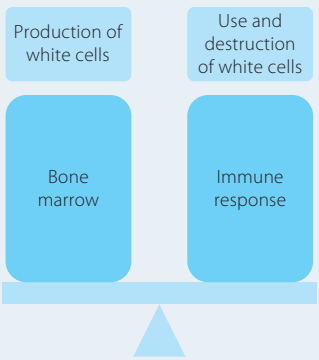
Bacteria	Neutrophilia
Viruses	Leukopaenia
Fungi	Neutrophilia; monocytosis
Protozoa	Eosinophilia; altered red cell architecture; increased IgM and IgG
Macroparasites	Eosinophilia; increased IgE

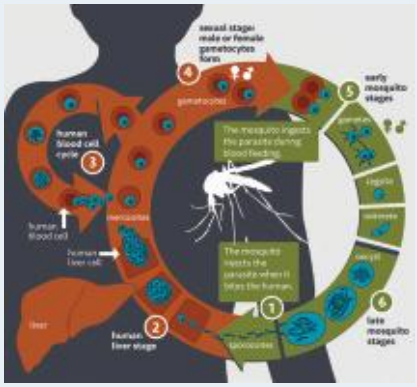


Using models to aid understanding

Scientific models

- Useful to represent complex ideas or objects, or objects that are very large or small
- Predictive of some aspect of the object or idea
- Modified as knowledge increases
- e.g. double helix model of DNA, heliocentric model of the solar system





12 CHAPTER REVIEW QUESTIONS

Review quiz

1. 'The innate immune system includes those responses to pathogens that are genetically programmed and available from birth.' Is this statement true or false?
2. Identify the four main features of the innate immune system.
3. Briefly distinguish between the two arms of the adaptive immune system.
4. Describe in detail the cellular and chemical mechanisms that allow the adaptive immune system to respond to specific pathogens.
5. Draw the structure of an antibody. Label the antigen-binding site and the light and heavy chains.
6. Explain the aspects of the adaptive immune response that allow it to respond to a wide diversity of pathogens upon primary exposure.
7. What is the difference between a primary response and a secondary response to a pathogen?
8. Evaluate the role of B and T memory cells in the adaptive immune response.
9. Describe the roles of antibodies in the response to pathogens.
10. What mechanisms allow T cells to respond to pathogen-infected cells? Use a flow chart to summarise the processes involved in identification and destruction of intracellular pathogens.
11. Create a table to summarise the roles of the following cells:
    - **a** helper T cells
    - **b** cytotoxic T cells
    - **c** suppressor T cells
    - **d** memory T cells.
12. Outline the role of MHC class I and MHC class II in the response to pathogens.
13. Evaluate the role of the light microscope when examining the tissues and cells involved in the innate and adaptive immune responses.
14. What are some of the features of a good scientific model? Use an example you have studied in class.
15. Assess the role of peer feedback when conducting a practical or secondary sources task. Why is it important for scientists to peer review each other's work?
16. Explain the processes that affect the balance of different types of white cells in the blood. How might this help a doctor to diagnose an infectious process?
17. Evaluate the role of full blood counts in the assessment of a patient presenting with symptoms of an infection.
18. Compare the roles of the humoral and cell-mediated immune responses with regard to the type of pathogen targeted and how pathogen destruction is brought about.
19. Compare and contrast the MHC class I and MHC class II molecules.
20. The adaptive immune system is sometimes described as more 'sophisticated' or 'important' than the innate immune system. Evaluate whether either or both of these adjectives fits.
21. Draw a diagram that shows all the different defences encountered by an antigen when it enters the body. Be sure to indicate how these different defences communicate.

Exam preparation

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13 Prevention, treatment and control of disease

INQUIRY QUESTION

How can the spread of infectious diseases be controlled?

Students:

- investigate and analyse the wide range of interrelated factors involved in limiting local, regional and global spread of a named infectious disease EU IU
- investigate procedures that can be employed to prevent the spread of disease, including but not limited to: (ACSBL124) CCT EU ICT IU
  - hygiene practices
  - quarantine
  - vaccination, including passive and active immunity (ACSBL100, ACSBL123) EU ICT
  - public health campaigns
  - use of pesticides
  - genetic engineering
- investigate and assess the effectiveness of pharmaceuticals as treatment strategies for the control of infectious disease, for example: CCT EU ICT IU
  - antivirals
  - antibiotics
- investigate and evaluate environmental management and quarantine methods used to control an epidemic or pandemic CCT IU
- interpret data relating to the incidence and prevalence of infectious disease in populations, for example: ICT N
  - mobility of individuals and the proportion that are immune or immunised (ACSBL124, ACSBL125)
  - Malaria or Dengue Fever in South East Asia AAEA
- evaluate historical, culturally diverse and current strategies to predict and control the spread of disease (ACSBL125) ATSIHC AAEA CCT IU L
- investigate the contemporary application of Aboriginal protocols in the development of particular medicines and biological materials in Australia and how recognition and protection of Indigenous cultural and intellectual property is important, for example: ATSIHC S
  - bush medicine
  - smoke bush in Western Australia

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
13.1 Processing data and information on the factors limiting spread of disease
13.2a Food safety standards
13.2b Analysing quantitative data from the 2010 Haitian earthquake
13.2c Evaluating scientific claims
13.3 The effect of introducing penicillin on death rates from infections
13.4 Investigating the incidence and prevalence of malaria in Cambodia
13.5 Historic control of a disease outbreak

Worksheets
- GM livestock and public concerns about genetically modified organisms
- Limiting the spread of an infectious disease - polio
- Controlling an epidemic or pandemic
- Preventing the spread of an infectious disease - polio
- Preventing mosquito-borne diseases by using vector control

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**


Dimas Ardian/Getty Images

FIGURE 13.1 A temperature monitor at Inchoen International Airport in Seoul, South Korea

The increased mobility of humans due to air and sea travel has had a profound impact on our ability to limit the spread of infectious disease. Regular and inexpensive international travel has allowed a greater proportion of the world's population to holiday or work overseas. When people travel, they take their pathogens with them. People who may be carrying an infectious disease then expose new populations to the pathogen. Some airports, such as those in Hong Kong or Seoul, use temperature-sensing equipment to screen people as they move through the arrivals gate, to detect possible carriers of diseases such as influenza (Fig. 13.1).

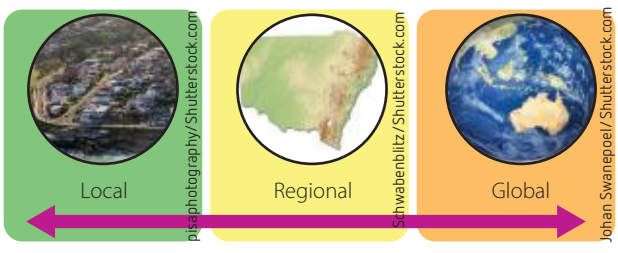
13.1 Limiting the spread of infectious disease

Applying the scientific method to the understanding of infectious disease avoids a lot of wasted energy pursuing the wrong course of action, as well as reducing panic.

Factors involved in monitoring and control

Because of the ease with which humans can travel, disease monitoring and control is carried out at three levels: local, regional and global (Fig. 13.2).

FIGURE 13.2 The three levels of disease monitoring and control



Local factors

Local factors are usually related to a neighbourhood, village, town or city. Major factors that influence the spread of disease include sanitation, especially how waste and sewage are disposed of. This is especially important after typhoons and hurricanes, where the sewerage system is disrupted. Overcrowding

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increases the chance of host-to-host transmission. Poor communication networks and roads may limit access to medical treatment, hospitals or even medical information.

Animal husbandry practices, such as keeping chickens and pigs, may facilitate the transmission of avian and swine flu from animals to humans. Keeping horses in south-eastern Queensland in the same area as wild fruit and insectivorous bats has led to the emergence of Hendra and Australian bat lyssavirus.

Local cultural and spiritual beliefs may influence attitudes to medical advice, burial rituals and suspicion towards Western medical practices. In Madagascar, for example, a traditional ceremony known as *famadihana* involves exhuming dead relatives and dancing with them through the streets. Health officials believe that this has contributed to a deadly outbreak of plague.

Intercultural understanding

Regional factors

The United Nations divides the world into five regions: Africa, the Americas, Asia, Europe and Oceania (Australia, New Zealand, Melanesia, Micronesia and Polynesia). The geography of a region influences disease transmission. A region may be characterised by mountains, deserts, rainforests or grasslands, and these geographical factors determine whether a population in that region is highly mobile or relatively isolated. People living in mountainous regions or on islands have a natural isolation factor and this reduces their chances of being exposed to infected individuals.

Around 60% of the world's population live in coastal regions. Bacteria and viruses reside in sea water and seafood. For example, hepatitis A virus may be concentrated in molluscs, such as oysters and clams. The source of these microbes is sewage disposal. This is a huge challenge to infectious disease control in regions such as South-East Asia, which have high coastal populations who rely on seafood as a source of protein.

Increased trade of fresh food around regional areas creates a possible source of pathogen transmission. For example, faecal contamination of frozen mixed berries imported from China in 2016 contributed to a number of Australians testing positive for hepatitis A.

Local seasonal variations in temperature and precipitation patterns may influence the availability of **vectors**. Monsoon-related infectious disease outbreaks such as cholera, typhoid, malaria and leptospirosis have been observed in equatorial regions.

Global factors

The increased movement of people around the globe due to travel and work (page 446) and migration of refugee populations also introduces difficulties in limiting the spread of infectious disease. Many refugees have experienced trauma, food insecurity, overcrowding and lack of access to basic health care, such as vaccinations. Pre-migration medical examinations are carried out to exclude such infectious diseases as tuberculosis, measles, malaria and polio.

Intercultural understanding

Misuse of antibiotics and other antimicrobial medications has led to a rise in resistant bacteria. This is a global threat to infectious disease control. Many strains of tuberculosis are now resistant to the antibiotics traditionally used to treat this disease.

An important factor that has arisen in the control of infectious disease is the ease of communication afforded by the Internet. It is possible to transmit accurate and up-to-date data on disease outbreaks as they occur. Communication between scientists is of vital importance in the control of infectious disease.

FIGURE 13.3 Four factors affecting infectious disease outbreaks

Factors involved in disease transmission

When there is an outbreak of an infectious disease, there is usually not one single cause. The causes are **multifactorial** (Fig. 13.3), involving local, regional and global factors. Only when the factors affecting transmission are understood can effective strategies to limit the spread of infectious diseases be implemented.

Pathogen factors

Each pathogen is different. Some are virulent and can cause disease even when present in low numbers, while others only do so in large numbers. Some pathogens form **natural reservoirs** in food, water and the environment. Others are not environmentally resilient and must be transferred directly from host to host. The **incubation period** may be short or long. Some pathogens are easily neutralised by water and disinfectants, whereas others are more resilient. The strategies used by pathogens to cause disease are called **virulence factors**.

Once the biology of the pathogen is understood, control strategies can be put in place to target the specific features of the pathogen.

Host factors

Just because a host is exposed to a pathogen does not automatically mean they will succumb to the disease. The human immune system has a number of barriers that for the most part are effective at dealing with challenges by pathogens (Fig. 13.4). There are a number of reasons why these barriers may not be effective and the pathogen continues to invade the host. Any **concurrent** illness in the host may reduce the effectiveness of the host's defence systems. For instance, cancer or HIV/AIDS patients and those with diabetes may be less resistant to another pathogen. Malnutrition can put a strain on the ability of a host to avoid infection. People in a developing nation with ongoing food shortages due to war, drought or poverty may be unable to avoid infection. The use of certain pharmaceuticals, such as corticosteroids or anticancer medications, may lower the body's barriers against pathogens.

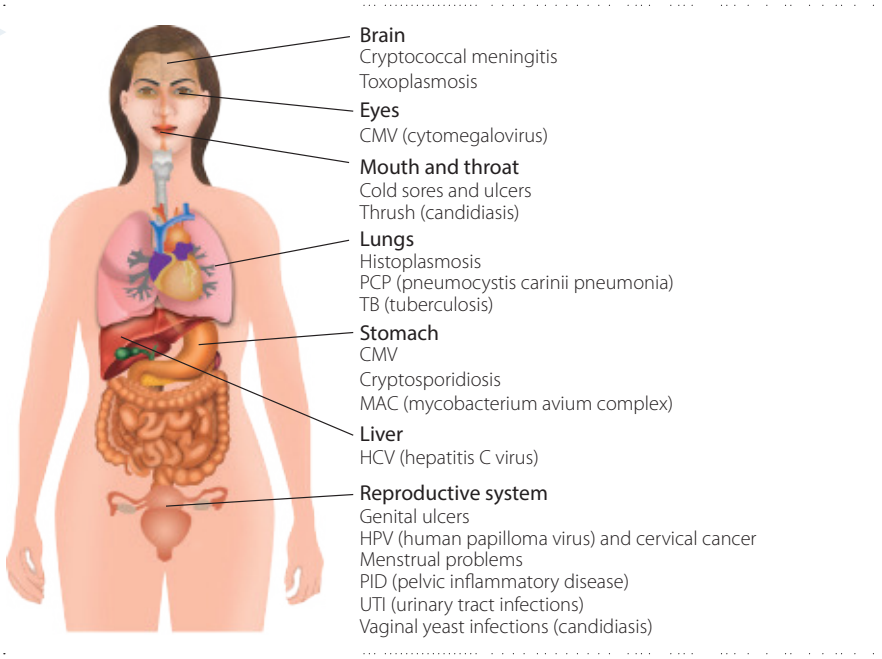

FIGURE 13.4 Opportunistic infections are more likely when the immune system is weakened.

Images & Stories/Getty Stock Photo

FIGURE 13.5 Typhoon Haiyan: a breakdown in infrastructure increases the risk of water-borne bacterial diseases.

Environmental and geographic factors

Certain environments may predispose to the spread of infectious disease. Wherever a pathogen is able to build up a large reservoir in the environment, there is a greater risk of outbreak. For instance, natural disasters such as earthquakes, hurricanes and volcanic eruptions may lead to poor sanitation and an increase in cholera-causing bacteria in local waterways (Fig. 13.5). Environmental conditions may favour the preservation of the pathogen in the environment. This is especially so in countries affected by malaria. For instance, mosquitoes require warm weather and a water body to reproduce and transmit the malaria parasite. Large-scale destruction of rainforests may expose reservoirs of hitherto unknown pathogens and put at risk those who use the land for housing and agriculture.

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INVESTIGATION 13.1

Processing data and information on the factors limiting spread of disease

During late 2017, the south-eastern United States and the Caribbean were devastated by a series of hurricanes, which caused widespread damage to infrastructure including water and power supplies, roads, homes and agricultural crops. Many communities, particularly in Puerto Rico, were isolated from the outside world for weeks. Despite this, the death rate from water-borne infections such as cholera was relatively low. After the Haitian earthquake of 2010, however, the story was very different – thousands of people died from cholera and other infectious diseases.

In this activity, you will work in a group of three to compile a management plan for a region that has experienced a natural disaster, incorporating the four factors affecting the spread of disease.

Critical and creative thinking

Intercultural understanding

Personal and social capability

Information and communication technology capability

AIMS

1. To select qualitative and quantitative data and information on management of the spread of an infectious disease using a range of formats
2. To apply quantitative processes to the analysis of infectious disease control
3. To evaluate and improve the quality of data obtained in your research

METHOD

1. With your group, brainstorm all the effects of a hurricane on communities. Include effects on both the natural and built environments.
2. Use your list to develop a series of immediate and long-term challenges to public health that could arise from the effects of a hurricane. These may include wound- and non-wound-associated infections.
3. Use online, printed resources and the weblinks to the right to gather the following quantitative and qualitative data on the spread of infectious disease during these two natural disasters (2010 and 2017):
   - a the incidence and mortality rates from cholera and other infectious diseases
   - b the incidence and death rate from wound infections
   - c a map of the area of devastation
   - d the populations of people affected
   - e the socio-economic factors affecting Haiti compared with the south-eastern United States (levels of poverty, health care available, median wages, levels of unemployment)
   - f the quality of the built environment, in terms of building design and materials, water and power supply, for both regions and its effect on mortality rates.
4. Examine the data you have gathered. Organise it in a way that allows you to make a comparison of the four factors involved in disease spread.

Weblink
Epidemics after natural disasters

Weblink
2016 Hurricane Matthew

Weblink
Communicable diseases following natural disasters

Weblink
Why disease hits harder after disasters in developing nations

RESULTS

Present your findings using an appropriate format as agreed with your teacher. Include a complete list of references.

DISCUSSION

1. Write an evaluation of the main factors that led to the vast difference in infectious disease rates in Haiti in 2010 compared with the United States in 2017.
2. Make recommendations based on your evaluation. What needs to change in the management of natural disasters in developing nations?
3. Evaluate the accuracy, reliability and validity of your data. Which sources of information were best at providing data that could be used to make a judgement?

CONCLUSION

Summarise some of the factors that increase the risk of infectious disease spread after a natural disaster.

Ethical understanding

Other societal factors

In Australia, preventable diseases such as chickenpox and measles are making a comeback. Anti-vaccination campaigns may influence people in deciding whether to have themselves and their children vaccinated. Lack of education regarding the factors involved in an epidemic can contribute to poor decision-making on the part of the public. Poverty and lack of access to vaccines and education is a factor in more remote communities.

The advent of mass human population movements due to armed conflict is a phenomenon that is gaining pace in the early 21st century. An unprecedented number of people are fleeing conflict in Africa and the Middle East, with many travelling thousands of kilometres from their homes. These people are at risk of malnutrition, exposure to the elements and exhaustion, all of which contribute to a lowering of their resistance to invading pathogens. While in transit, refugees stay in camps, where the hygiene of the food and water is questionable. Overcrowding increases the likelihood of host-to-host transmission, with the very young and very old most vulnerable. In cases where refugees are fleeing over a large distance, they may pass through transit zones where there is a greater exposure to pathogens due to the environmental conditions. There is evidence that the choice of route between origin and destination has affected the incidence of infectious diseases in refugees arriving in camps.

Isolated societies with small gene pools may be at greatest risk of disease outbreaks. A lack of variation in innate immunity and the absence of adaptive immunity can be devastating for a small and isolated population. When Aboriginal Australians were first exposed to European diseases such as smallpox, many succumbed due to a lack of previous exposure.

The rise of international travel by air and sea means an increase in the likelihood of transmission of disease outbreaks. A person carrying a pathogen as an air passenger on a long-haul flight is contained in a small space with a large number of people in close proximity, all sharing an air supply.

KEY CONCEPTS

- There are a wide range of factors involved in limiting the spread of disease and these can be identified at a local, regional and global scale.
- The causes of disease transmission are multifactorial and involve pathogen, host, environmental/geographical and societal factors.

CHECK YOUR UNDERSTANDING 13.1

1. Explain why disease transmission needs to be dealt with at local, regional and global levels.
2. List the four main factors involved in infectious disease outbreaks and provide one example of each.
3. List factors about the pathogen that affect the rate and extent of disease transmission.
4. List reasons for the differences in infectious disease prevalence in different societies. What role do culture and education play in the spread of infectious disease?
5. Justify why the correct management of an environment alone could have the potential to eradicate infectious disease in an area. Use an example.

13.2 Preventing the spread of infectious disease

Throughout history, humans have been investigating ways in which the spread of disease can be prevented. Over 3000 years ago, the Chinese and Hebrews, along with some other cultures, were advocating cleanliness in food, water and personal hygiene. The Hebrews had rules to be followed to ensure that the health of their population was maintained. Some of these rules included washing people and objects, proper disposal of refuse and excreta, protection of the water and food supply, and isolation of diseased individuals. For example, people suffering from leprosy were forced to live outside the walls of the city. Principles of good health were also developed by certain groups of the Chinese population

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and included hygiene rules and exercises. Similarly, the people of Mesopotamia followed principles of cleanliness, used toilets and constructed primitive sewerage systems.

While many cultures have practised the principles of good health, infectious diseases such as bubonic plague and cholera have caused widespread death over many years.

Hygiene

Hygiene can be divided into two types: personal and community hygiene.

Personal hygiene involves each person keeping their body and any openings on it clean. This reduces the risk of pathogens entering our bodies, or transmission of these pathogens to others, thus causing disease. It also inhibits the build-up of micro-organisms on our bodies. Personal hygiene includes the following practices:

- Hands should always be washed with soap and water before preparing and eating food and after going to the toilet (Fig. 13.6). This prevents the spread of pathogens that cause symptoms such as diarrhoea.
- The body and hair should be washed regularly and teeth cleaned, to prevent the build-up of pathogens (particularly bacteria) to numbers sufficient to cause disease. For example, a build-up of bacteria in the mouth can cause gingivitis (resulting in swollen, bleeding gums and loss of teeth).
- Always cough or sneeze into a handkerchief or tissue. This prevents airborne droplets from spreading to others. You should not sneeze into your hands, as you will then transmit the pathogens from your hands to whatever you touch next.

The Australian Institute of Food Safety

FIGURE 13.6 The Australian Institute of Food Safety's 'A guide to washing your hands' poster

Community hygiene helps prevent the build-up of pathogenic organisms in the community. When the infrastructure that supports and maintains community hygiene breaks down, there is a rapid spread of disease, as was seen in the aftermath of the tsunami that hit South-East Asia in 2005.

Community hygiene includes the following measures:

- Sewage and garbage disposal reduce the risk of pathogen numbers increasing and spreading throughout the community. Sewage treatment plants may incorporate the use of UV disinfection for management of pathogens in waste water.

FIGURE 13.7 Overcrowded cities can put a strain on water and water management, leading to poor disease control.

- Sterilisation and disinfection of equipment in hospitals, doctors' surgeries, dentists and hairdressers/barbers reduces the risk of the spread of pathogens from one person to another.
- City planning reduces overcrowding (Fig. 13.7) and therefore reduces the risk of the transmission of diseases throughout the population – this is important in controlling disease.

Clean food and water

Many pathogens can be transferred from person to person, or from environment to person, in food. For example, salmonellosis is a disease caused by *Salmonella* bacteria and is transmitted in undercooked food, especially of animal origin. In order to control the transmission of disease from food sources, guidelines that must be followed by food handlers have been introduced for storing, preparing and serving food.

It is important that water quality is maintained in order to minimise the risk of pathogens multiplying and to reduce the risk of the transmission of pathogens in contaminated water.

Domestic water quality must comply with strict standards and guidelines in order to reduce the incidence of disease. In Australia, governments establish these standards and the water is tested daily to ensure these standards are met.

Water that has been contaminated with the faeces of animals could contain unsafe levels of pathogens, such as the protozoans *Cryptosporidium* and *Giardia*, and if consumed could cause symptoms such as abdominal cramps, diarrhoea, nausea and vomiting. Cholera is a potentially fatal disease that is transmitted in water contaminated with untreated sewage.

The treatment of water to destroy pathogens and prevent their further multiplication reduces the transmission of disease and is very important in the successful control of disease.

INVESTIGATION 13.2a

Food safety standards

Work and enterprise

Civics and citizenship

Weblink
Food Standards Australia and New Zealand

Weblink
Food safety fact sheet

Food safety standards are enforceable guidelines for Australian food businesses. The aim of these standards is to minimise the spread of pathogens from contaminated food and water, which could cause ill health and/or death.

AIM

To research and assess the Australian Food Safety Standards, including guidelines for:

- personal hygiene practices
- maintenance of utensils used for cooking
- storage of raw and cooked foodstuffs
- processing (meat)
- preparation and cooking methods

METHOD

1. Use online and printed sources to examine the Australian Food Safety standards, to gather information on the above aspects of food safety. Concentrate on those aspects of the guidelines that are there to limit the spread of pathogens and therefore control disease outbreaks.
2. Gather your data from reliable, accurate and valid sources.

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RESULTS

Present your findings using an appropriate format as agreed with your teacher. Include a complete list of references.

DISCUSSION

Outbreaks of infectious disease still occur in commercial premises despite the laws and guidelines. Suggest reasons why contamination from pathogens in food will always be a risk despite our advanced scientific understanding of the control of infectious disease.

Weblink
Department of Primary Industries – Food Authority

INVESTIGATION 13.2b

Analysing quantitative data from the 2010 Haitian earthquake

When natural disasters such as earthquakes occur, they generally have a more devastating effect where there is a greater population density. Management during and after a natural disaster is complicated by economic, social and cultural factors. Although many deaths are a direct result of collapsed buildings, a great proportion of deaths are attributable to infectious disease spread after the disaster. In this activity, you will analyse data regarding the 2010 Haitian earthquake and use this to make informed judgements about the factors that affect the spread of infectious disease after a natural disaster.

Critical and creative thinking

Numeracy

Information and communication technology capability

AIMS

1. To derive trends, patterns and relationships in data and information
2. To assess the relevance, accuracy, validity and reliability of data

METHOD

1. Examine Figure 13.8. What is the general trend in the data?

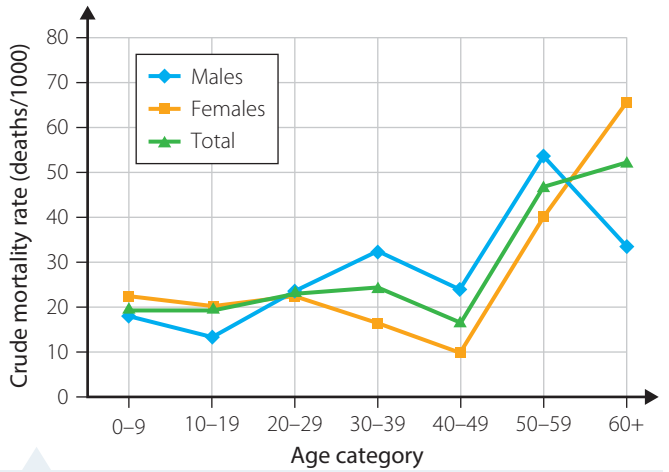

FIGURE 13.8 Age- and sex-specific mortality rates following the 2010 Haitian earthquake

2 Complete the final column in Table 13.1. Identify any trends in the data.

3 Identify possible factors that could have affected the death rates of males and females.

TABLE 13.1 Mortality estimates from the 2010 Haitian earthquake, according to individual and household characteristics

	TOTAL EXPOSED	TOTAL DEATHS	MORTALITY RATE (%)
Overall	6383	153	
Sex			
Male	3052	71	
Female	3379	76	
Age category			
0–17	2299	37	
18–49	3689	92	
50+	548	24	
Education level			
None	289	11	
Primary	1237	27	
Secondary	3685	83	
Higher education	1273	31	
Crowding			
<2.0	1927	35	
2.0–2.9	1566	37	
3.0–3.9	949	23	
4.0+	2094	58	
Current location			
Neighbourhood	3258	63	
Camp	3278	90	
Multilevel building			
1 level	3999	76	
>1 level	2031	67	

Source: adapted from Doocy S, Cherewick M & Kirsch T 2013, 'Mortality following the Haitian earthquake of 2010: a stratified cluster survey', Table 2, https://pophealthmetrics.biomedcentral.com/articles/10.1186/1478-7954-11-5

DISCUSSION

1 The spread of infectious disease is affected by four main factors (page 433). Using each of these factors, explain the trends in the data in Table 13.1 and Figure 13.8.

2 Assess the accuracy of the data in Figure 13.8. What factors do you think determine the ability of scientists to collect accurate population and demographic data in countries such as Haiti?

3 Explain why accurate data in natural disasters such as this earthquake is vital in the management of infectious disease spread.

4 What other individual or household factors in Table 13.1 would assist our understanding of the spread of disease?

CONCLUSION

From your analysis of the data provided, which individuals were most likely to die from disease after the earthquake in Haiti in 2010?

KEY CONCEPTS

- The spread of disease may be limited by a combination of strategies, including hygiene practices around food and water.
- Personal and community hygiene are designed to limit the transfer of pathogens from person to person.
- Food and water cleanliness are designed to control the outbreak of disease by reducing the pool of pathogens in food and the environment.
- During natural disasters, many of the factors affecting the spread of disease come into play all at once. This can cause a massive increase in the spread of infectious diseases.

CHECK YOUR UNDERSTANDING 13.2a

1. Australia has some of the strictest food and water hygiene practices in the world. Justify the cost (in time and personnel) of the practices put in place to ensure adherence to the recommended guidelines.
2. Scientists have a very good understanding of the causes and management of food and water-borne diseases. Why, then, do we still hear in the media about incidents of food poisoning in Australia despite our knowledge and technology?
3. Imagine you have completed a first-hand investigation to identify microbes in food or in water. Assess how the equipment you used would be useful in the field during a disease outbreak. Which aspects of the investigation would be the most challenging? List the precautions you would take to make sure that your surroundings and equipment were sterile when completing the investigation. Justify these precautions.
4. Explain how each of the following assists in the control of disease in a fast-food establishment:
   a. personal hygiene practices
   b. food practices
   c. water practices.
5. Describe the processes involved in the treatment of water to ensure that it meets the guidelines issued by the National Health and Medical Research Council to make it safe for drinking. Explain how these processes reduce the risk of infection from pathogens.

Quarantine

Australia is one of few countries in the world that remain free of the world's most serious pests and diseases. Originally, this was due mainly to our geographical isolation. This isolation decreased as international travel and trade increased, which led to the need for a more sophisticated and thorough system to prevent the entry of pests and diseases into our country. The Department of Agriculture and Water Resources (DAWR) is responsible for maintaining Australia's reputation as a relatively disease-free country.

Australia is home to many unique flora and fauna species, as you will know from the Year 11 Biology course. Australia also has a very large agricultural industry that supplies large amounts of food for local consumption and export (worth $30 billion per year). Our agricultural exports are in high demand in many countries because of Australia's reputation of being free of the serious pests and diseases that are common in other areas. If these pests and diseases were to gain entry into Australia, they would have a devastating effect on Australia's animals and plants.

The role of **quarantine** is to minimise the risk of exotic pests and diseases entering Australia, in order to protect our native flora and fauna, our agricultural industries, our environment and our health.

Australia's thousands of kilometres of coastline, and proximity to neighbouring regions such as South-East Asia and the Pacific nations, which have pests and diseases that are not present in Australia, make it particularly vulnerable to pest and disease invasions. These pests and diseases could be brought into Australia by people, animals and plants. They could also be brought in by animal and plant products, or in soil that is on shoes or machinery.

Animal quarantine

Animal quarantine involves all animals that come into Australia spending time at quarantine stations, to ensure that they are free of disease before they are released. If you wanted to bring pets into Australia, you would have to leave them in quarantine for a number of weeks. These animals would be examined regularly for signs of disease.

Plant quarantine

Plant quarantine involves examining all plants, parts of plants and plant products (fruits, seeds, cuttings, bulbs and wood) brought into Australia, for pests or diseases. Many of these items are refused entry into Australia. In some cases the items are allowed into Australia only after being treated by quarantine officers, to ensure that any likely pests or pathogens are destroyed. Live plants must be kept at quarantine stations until any diseases that may be present have had time to develop.

Human quarantine

The captains of aircraft and ships are required to notify Australian Quarantine and Inspection Services (AQIS) if any passengers or crew are displaying any symptoms of prohibited diseases, such as rabies, yellow fever, malaria, SARS (severe acute respiratory syndrome) or avian influenza (bird flu). Aircraft are also sprayed with insecticide to kill any pests that have entered Australia with the aircraft. All Australian international airports have mosquito-trapping programs to enable the quick detection of any mosquitoes entering the country. These mosquitoes could be acting as vectors in the transmission of disease.

Northern Australia Quarantine Strategy

The northern part of Australia is only a short distance from countries that have many exotic pests and diseases not present in Australia. The Northern Australia Quarantine Strategy (NAQS) was established as an early warning system to protect this susceptible area of Australia. This system involves the use of 'sentinel' animals such as cattle and pigs that are regularly checked for diseases, such as Japanese encephalitis and blue tongue. Sentinel animals are so called because they are in effect 'standing guard' or 'watching out' for invading pathogens or pests. If they develop symptoms of a disease not yet seen in Australia, it is an early warning sign that these pathogens are a threat to Australia, and strategies can then be put in place to halt their spread. Insect traps are also set up and checked regularly for insects such as screw-worm flies, Asian honeybees and papaya fruit-flies.

The DAWR also has many roles in keeping Australia free of infectious diseases (Fig. 13.9).

Vaccination

You learned about the primary and secondary immune responses in Chapter 12.

Vaccination involves the introduction of a vaccine into the body. **Immunisation** is the process in which the body reacts to a vaccine by going through the immune response. This response produces memory cells for the antigen and confers immunity to the body, so that if the antigen enters the body again in the future, the secondary response will occur and the person will avoid the worst symptoms of the disease.

Vaccination primes the immune system to deal with a pathogen it has never been in contact with. There are several important reasons why we don't just let the body experience the disease and build up a natural immunity.

Active acquired immunity is when the immune response occurs and memory cells are produced. It can be naturally induced, as the body undergoes the immune response and suffers the symptoms of the disease. It can also be artificially induced, through the use of vaccines, which cause the production of memory cells without the body experiencing the symptoms of the disease.

Vaccines contain cultures of micro-organisms, which may be either:

- living but attenuated (weakened) and therefore harmless (rabies, poliomyelitis, measles), or
- dead (typhoid, whooping cough).

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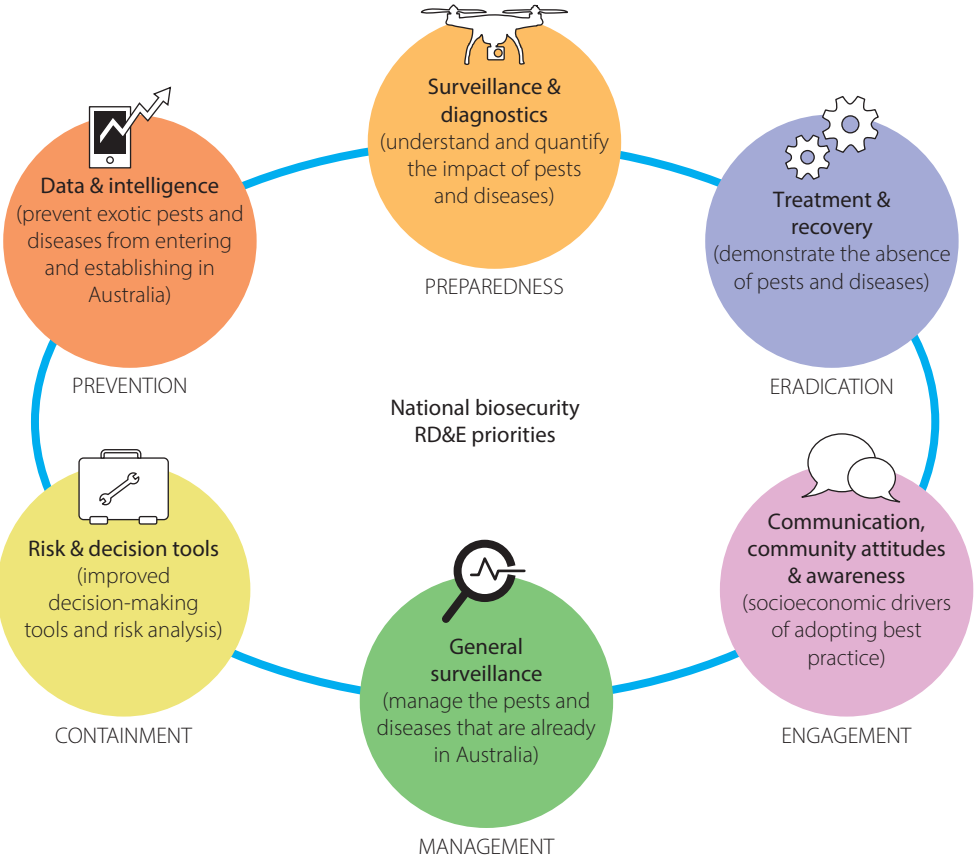

FIGURE 13.9 DAWR has many roles in keeping infectious diseases out of Australia (RD&E: research, development and extension).

Vaccines may also contain modified toxins called toxoids (tetanus, diphtheria). They may be given orally, by injection or by scratching the skin surface.

Vaccines are harmless to the body and will not cause the disease that they are specific for. They contain the antigens that cause the body to undergo an immune response and produce memory cells for that particular antigen. If the body is exposed to that antigen in the future, the secondary response will be activated and the antigen will be destroyed before any symptoms of the disease are experienced. The immunity formed in this way is usually lifelong. Each vaccine is specific for only one type of antigen and will therefore give immunity for only one type of disease.

For a vaccine to be effective, a series of vaccinations (booster shots) should be given over a number of years. Each time the vaccine is introduced into the body, a small response is produced. Over a series of vaccinations, the lymphocytes will more rapidly recognise the antigen and the numbers of memory cells produced will be enough to give immunity for a long time.

In some cases, the numbers of memory cells decrease over time and booster injections have to be given to increase the number of circulating memory cells, to ensure that immunity is maintained for that disease. For example, booster injections must be given to maintain immunity to tetanus, as the number of memory cells for this disease decreases over time.

Passive acquired immunity involves the introduction of antibodies (immunoglobulins) into the body to prevent a disease from developing. These antibodies have been produced by another organism that has had the disease. For example, if you have been exposed to hepatitis A, you may be given injections of immunoglobulins (antibodies) to prevent you from contracting the disease. This immunity will last for only a couple of months, as no memory cells are produced.

Vaccines are a vital part of the solution to infectious disease control. Investigation 13.2c shows the effectiveness of vaccines in controlling a group of childhood infectious diseases.

INVESTIGATION 13.2c

Ethical understanding

Information and communication technology capability

Evaluating scientific claims

You will evaluate the effectiveness of a vaccination program in preventing the spread of diseases including smallpox, diphtheria and polio. You will evaluate the claims made by health authorities regarding the efficacy of vaccination programs.

AIM

To analyse and evaluate primary and secondary data and information on vaccination programs and their effectiveness in limiting the spread of infectious diseases

METHOD

Weblink
Immunise Australia Program

Task 1 Read the information at the *Immunise Australia Program* weblink and then answer the questions below.

1. Explain why it is necessary to vaccinate against polio.
2. Outline the vaccination schedule in Australia.

Weblink
Polio immunisation

Task 2 Read the information at the *Polio immunisation* weblink and then answer the questions below.

1. Is the current vaccine given in Australia the oral or injectable vaccine?
2. Are any of the side effects of the injectable polio vaccine long term (lasting several years or a lifetime) or life threatening?
3. Explain why it is important to eradicate polio globally.

Weblink
Better Health Channel: Polio immunisation

Task 3 Read the information at the *WHO poliomyelitis* weblink and then answer the questions below.

1. Outline the aims of the Global Polio Eradication program, which began in 1988 and was revised in 2012.
2. Polio is still endemic (present) in three countries: Afghanistan, Pakistan and Nigeria. There are also currently outbreaks in the Horn of Africa, western central Africa and Syria.

   Using information from the weblink, explain the science behind this statement:

   > We wouldn't have the outbreaks if we didn't have the polio virus continuing to circulate in the endemic countries.
   >
   > – P. Crowley, UNICEF ('The World Today' ABC Radio)

Weblink
WHO poliomyelitis

Task 4 Read the information at the *Polio outbreak sparked by vaccine* weblink and then answer the questions below.

1. Explain why people in certain countries believe that polio vaccination is a plot to sterilise them.
2. Find out whether science verifies or discredits this claim.

Weblink
Polio outbreak sparked by vaccine

Task 5 Go to the *Polio eradication* weblink and read the sections titled, 'Polio eradication basics' and 'Weakest link no. 1'. Then answer the question below.

1. Do scientific studies and explanations verify or discredit this claim in Nigeria? Justify your answer.

Weblink
Polio eradication

Task 6 Read the information at the *Progress towards eradication* weblink and then answer the questions below.

1. Discuss the change in **morbidity** (number of cases), **mortality** (% population deaths), **incidence** (number of new cases in a specific time) and **prevalence** (number affected at any one time) of polio from 1998 to 2014.
2. Describe how these figures are expected to change within 10 years if polio is not eradicated in these endemic countries.

Weblink
Progress towards eradication

DISCUSSION

1. State the World Health Organization's standpoint on polio vaccination. Do you agree or disagree with this standpoint? Justify your answer.
2. Analyse the reasons why a small proportion of the population remain deeply suspicious of vaccinations.
3. Evaluate the websites used in this activity in terms of their relevance, accuracy, validity and reliability.
4. Use online resources to examine and evaluate the claims made about a link between autism and vaccination.

CONCLUSION

Summarise the arguments in favour of vaccination as one strategy for limiting the spread of infectious disease.

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Public health campaigns

The scale and complexity of infectious diseases can seem overwhelming. We are confronted every day with news images of human suffering, particularly the rise of infectious diseases associated with the wave of human population movement that is taking place during the 21st century. However, any initially overwhelming task can be broken down into a series of smaller steps – you will be familiar with using this approach in completing large and complicated school assessment tasks.

The approach to controlling infectious diseases can be broken down into four categories, known as RICE (Fig. 13.10):

- *Resolution* of governments and health organisations (such as the World Health Organization) to find solutions to infectious disease
- *Information* in the form of epidemiological studies and scientific studies of the pathogen and its mode of transmission so that the solution is based on accurate data; includes increased national funding for scientific research/and training of scientists and medical personnel
- *Coordination* of efforts on a local, regional and global scale so that resources are used efficiently
- *Education* of human populations on a local, regional and global scale regarding factors affecting infectious disease transmission.

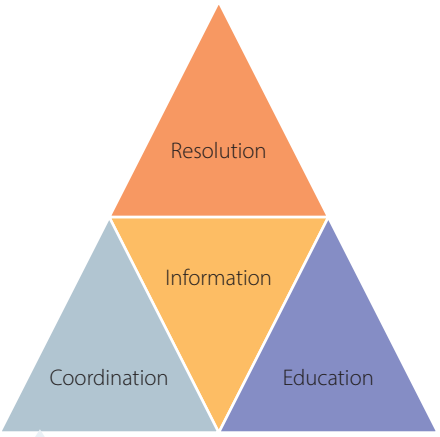

FIGURE 13.10 Three levels of approach to infectious disease limitation

There are a number of ways in which disease can be controlled and/or prevented. You have already read about two strategies: vaccination and quarantine. Another strategy for control and/or prevention of disease is the implementation of public health programs.

Government regulations ensure that standardised procedures are followed when handling, cooking and storing food. Strict guidelines must also be followed in hospitals, surgeries and clinics when sterilising equipment and when health workers move from patient to patient. When these guidelines and procedures are followed, the spread of pathogens is prevented; in turn this prevents the occurrence of diseases such as food poisoning and hepatitis. Government regulations are also in place to ensure that garbage is disposed of correctly, drinking water is treated effectively, and sewage is removed and treated. If these sanitation procedures are followed correctly, the spread of pathogens is prevented and, hence, the occurrence of disease in individuals and communities is prevented.

The law requires that certain diseases be reported to authorities if they are detected. This allows the early detection of these diseases and allows appropriate strategies to be put in place in order to control their spread through the community. Examples of **notifiable** diseases are measles, botulism, cholera, meningococcal infection, pertussis (whooping cough) and malaria.

Public immunisation programs (such as childhood immunisation) for diphtheria, tetanus, whooping cough, measles, mumps and rubella help to prevent these diseases. Mass immunisation programs for the human papilloma virus have also been introduced, to help prevent cervical cancer.

Use of pesticides

Pesticides are chemicals used to kill the pests of plants and animals, including pathogens and the vectors that transmit pathogens between organisms. Killing these pests and vectors reduces the occurrence of disease or controls the spread of disease through the population. Pesticides can be classified into three groups:

- insecticides – kill insects
- fungicides – kill fungal pathogens
- herbicides – kill weeds.

One of the best-known insecticides used to kill insects acting as vectors is DDT. It was used widely during World War II to controlled the spread of typhus. DDT killed the lice that transmitted the pathogen (bacteria) that caused the disease. DDT has also been used widely to kill the *Anopheles* mosquito, which carries the plasmodium that causes malaria. DDT controlled the spread of malaria, as transmission of the pathogen was prevented by the death of the vector. However, the effectiveness of DDT was reduced as mosquitoes developed resistance to it due to natural selection. Many countries have since banned the use of DDT because of its harmful effects on the environment. Some malaria-infested countries still use DDT to control mosquito numbers, even though it is not as effective. Other insecticides, such as pyrethrum, are now also used. These insecticides are less harmful to the environment and more effective in controlling mosquito numbers. This in turn controls the spread of malaria to some extent.

Another vector controlled by the use of insecticides is the aphid that carries the potato leaf-roll virus, which causes stunted growth and serious loss of yield in potato plants. Pesticides are used to kill the aphids and therefore control the spread of the virus that causes this devastating disease.

Pesticides are also used widely to spray items brought into Australia, to kill any insects or other organisms present. This prevents the spread of any diseases associated with these insects.

One problem associated with the use of pesticides is the ability of insect vectors and disease-causing organisms to build up resistance, which reduces the effectiveness of the pesticide and increases the need for the development and use of stronger pesticides.

The use of pesticides is also being discouraged further by their damaging effects on the environment. The use of natural pesticides is gaining popularity – these are developed from natural sources such as onions and garlic. In addition, companion plants may be grown in vegetable gardens. These plants are grown within a crop and are said to have insecticidal properties. They may even attract beneficial insects.

Genetic engineering

Ethical understanding

Genetic engineering involves altering the genetic composition of an organism. By altering the genetic make-up of organisms, it is possible to make them resistant to diseases. This prevents the disease occurring and controls the spread of disease through the population. Organisms with genes from other organisms inserted into their own genetic material are called transgenic species. (See Chapter 9, page 295.)

Weblink
Ethics of GM foods

Using genetic engineering to produce disease-resistant plants and animals prevents the occurrence of disease in individual organisms, controls the spread of disease through the population, and reduces the incidence of the particular diseases and pests.

Worksheet
GM livestock and public concerns about genetically modified organisms

The use of genetically modified organisms has not been universally accepted, however. There are concerns that resistance to the insecticides produced by the genetically altered organisms will develop and they will no longer be effective. There are also concerns about the effect these organisms will have on the environment and biodiversity. There are also many ethical issues to be considered, especially concerning public labelling of products derived from genetically modified organisms.

KEY CONCEPTS

- A wide range of measures can be employed to prevent the spread of infectious diseases.
- Hygiene practices prevent the creation of a pool of pathogens from which disease can spread.
- Quarantine practices limit the spread of pathogens by isolating infected hosts until the pathogen is eliminated, thus preventing transmission from host to host.
- Vaccination prepares the host for any future challenge from a pathogen.
- Public health programs raise levels of awareness and knowledge about the causes and transmission of infectious disease. This enables people to make more informed choices based on scientific knowledge.
- Pesticides are designed to kill pathogens directly or eliminate the vectors that transmit them.
- Genetic engineering is one option for creating organisms that are resistant to the effects of the pathogen without having to rely on costly vaccination programs.

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CHECK YOUR UNDERSTANDING
13.2b

1. What is the aim of quarantine in relation to disease prevention?
2. Provide two reasons why Australia is a difficult country to quarantine.
3. Australia's quarantine procedures are some of the strictest in the world. Justify the strict quarantine procedures employed for animals, plants and humans at airports and seaports in Australia.
4. Explain why booster shots are necessary.
5. Distinguish between active acquired immunity and passive acquired immunity.
6. Assess the effectiveness of the RICE approach to infectious disease limitation (Fig. 13.10).
7. How would you respond to the claim that pesticides are effective in the short term for eliminating vectors but in the long term have serious negative consequences that outweigh the short-term benefits?
8. Evaluate the data in Figure 13.11. What types of variables would have to be controlled to draw a valid conclusion about death rates from infectious disease and the introduction of vaccination programs for various infectious diseases?

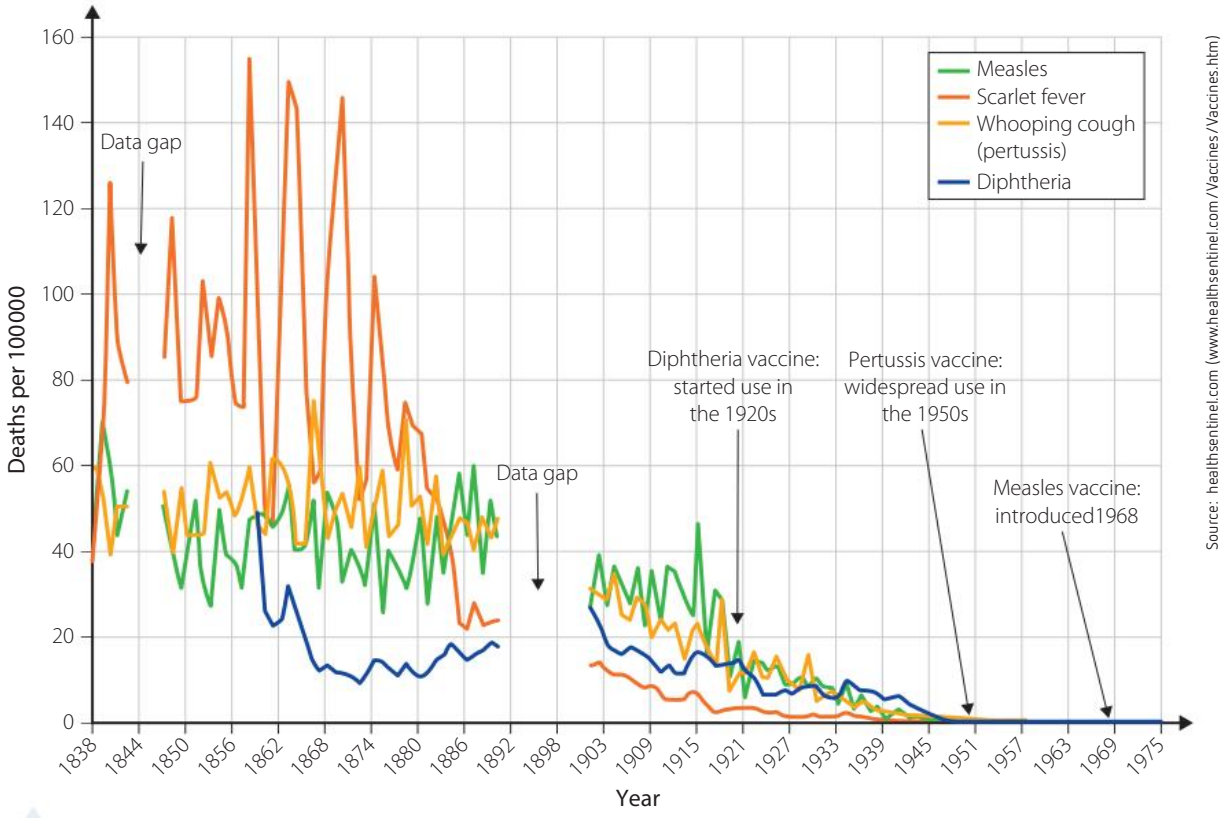

FIGURE 13.11 Mortality rates from infectious diseases in England and Wales before and after the introduction of vaccination

13.3 Pharmaceuticals for controlling infectious disease

The term **chemotherapy** has become very much associated with cancer treatment. However, in clinical terms, 'chemotherapy' means the use of any drug to treat any disease. The chemotherapeutic approach to infectious disease control involves the use of medications to control infectious diseases in humans, plants and animals. **Antimicrobial agents** are designed to control infectious diseases caused by microbes.

The main classes of antimicrobials are listed in Table 13.2.

TABLE 13.2 Classes of antimicrobial drugs

CLASS OF MEDICATION	PATHOGENS TARGETED	EXAMPLES
Antibiotics (antibacterials)	Bacteria	Penicillins, tetracyclines, polymixins, sulfonamides, cephalosporins
Antivirals/ antiretrovirals	Viruses	• Tamiflu® (oseltamivir) used to treat influenza (antiviral) • Abacavir (antiretroviral used in HIV/AIDS treatment)
Antifungals	Fungi	Fluconazole, amphotericin B, caspofungin
Antiprotozoals	Protozoa	Doxycycline, metronidazole, mefloquine

Antiviral medications

Antiviral medications are used to control viral infections. They do not kill viruses, but inhibit their development inside infected cells. They do not cure the disease but simply slow down its progress, allowing the body's natural defences to take over. If taken early in the course of the disease, symptoms will be milder and of shorter duration. They stop the spread of viral diseases and therefore are a useful addition to the control of epidemics and pandemics.

Many people in the developing world may carry a number of viruses, including hepatitis B and C, and HIV. Management of these viral infections is complicated – simply dosing people with antiviral drugs does not ensure a cure. One of the major problems in the developing world is *access* to medications and *compliance* with dosing regimens. Many of these drugs need to be taken for months or for life.

Antivirals are a relatively recent development compared to antibiotics. The viruses most commonly targeted by antiviral drugs include:

- HIV (HIV is the virus; AIDS is the disease caused by the virus)
- seasonal influenza A
- herpes
- hepatitis B and C.

Viruses use the host's cells to produce new virus particles, making it very challenging to develop a class of drugs that stops viral replication without killing the host cells. With the rise of the HIV epidemic in the 1980s, there was a coordinated global medical research effort that led to great advances in our understanding of the biology of the virus.

The genetics of viruses vary in the following ways:

- They may contain DNA or RNA.
- The genetic material may be single-stranded (ss) or double-stranded (ds). For example, the hepatitis B virus is a dsDNA virus; HIV is a ssRNA virus.

Once the life cycle and genetics of viruses were better understood, scientists identified a number of possible target stages for antiviral drugs. Figure 13.12 shows the main stages in viral replication in a host cell. Combination therapy targeting different stages of the virus life cycle may enhance the effectiveness of antiviral management.

Three antiviral medicines to treat influenza are registered for use in Australia:

- oseltamivir (Tamiflu®)
- zanamivir (Relenza®)
- amantadine (Symmetrel®).

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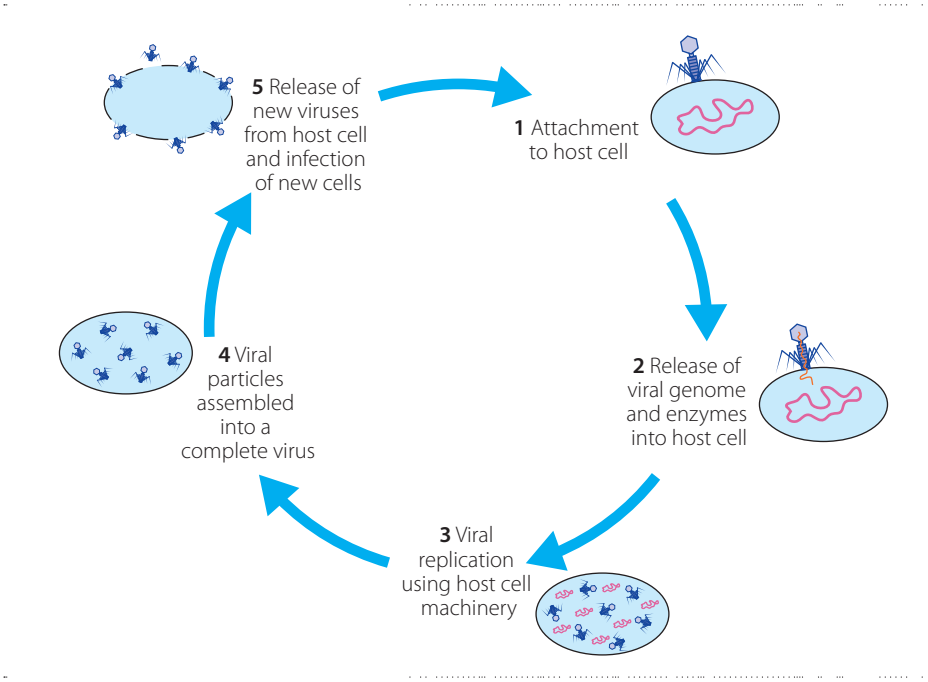

FIGURE 13.12
Stages in the replication of a virus in a host cell

These compounds are effective against seasonal influenza A strains, but scientists are still cautious about their effectiveness against a pandemic of influenza A subtype H1N1. There is evidence of beneficial effects in patients with lower respiratory complications, such as pneumonia. Reduced death rates were associated with the use of Tamiflu® and Relenza® during pandemics in 2009–2011. Further trials are needed. The question of **efficacy** is important, as nations stockpile these types of medications in case of a pandemic. The efficacy of a medication is its ability to produce the desired outcome. These drugs have greater efficacy when taken early in the course of the illness. There are concerns, however, about the safety of their widespread administration to populations during outbreaks.

Antibiotics

Antibiotics are used to control bacterial infections. They work by either killing or slowing down the growth of bacteria. Antibiotics are not effective against viruses.

Antibiotics are most effective when:

- they are used solely for the treatment of bacterial infections and not viral infections
- bactericidal antibiotics are used to kill rather than inhibit growth of the bacteria (penicillins, cephalosporins)
- narrow-spectrum antibiotics are chosen that target the specific pathogen
- they are able to get to the site of infection and kill the bacteria (the blood–brain barrier, necrotic and granulating tissue may be barriers)
- therapeutic blood levels are maintained
- the whole course is taken, to reduce the risk of bacterial resistance – although a recent scientific paper has questioned the validity of this central dogma of antibiotic treatment. Doctors will need to wait and see what subsequent studies may find before they change this practice
- a Gram stain and culture and sensitivity tests are done, to ensure that the appropriate antibiotic has been chosen and that the bacterium has been correctly identified as the causal agent of the disease.

INVESTIGATION 13.3

The effect of introducing penicillin on death rates from infections

Information and communication technology capability

Antibiotics selectively kill or inhibit bacteria. In this way, they contribute to the control of infectious disease by reducing the pool of living bacteria available in hosts. Antibiotics have no efficacy against viruses. The discovery of penicillin by Alexander Fleming in 1928 and the subsequent commercial development of antibiotics has had a dramatic effect on death rates from bacterial infections. This is most notable in the reduction in infant mortality and **perinatal** deaths of mothers.

METHOD

Use the data in Figure 13.13 to analyse the relationship between death rates from infectious diseases and the introduction of penicillin (an antibiotic) in the early 1940s.

RESULTS

1. Justify the addition of chlorine to drinking water, based on the information in Figure 13.13. How does this reduce the death rate from infectious disease?
2. What is the general trend in the graph over the 20th century?
3. What effect did the introduction of penicillin have on death rates from infectious diseases?
4. What was happening in the world at the time that penicillin was introduced? Comment on the 'good timing' of its introduction.
5. Use online secondary resources to gather information about the first introduction of penicillin.
   - a. How was it administered (tablet, injection or other)?
   - b. What effects did doctors report, especially those who were field medics in the army during WWII?
   - c. How is penicillin made? What is the source of the active ingredient?

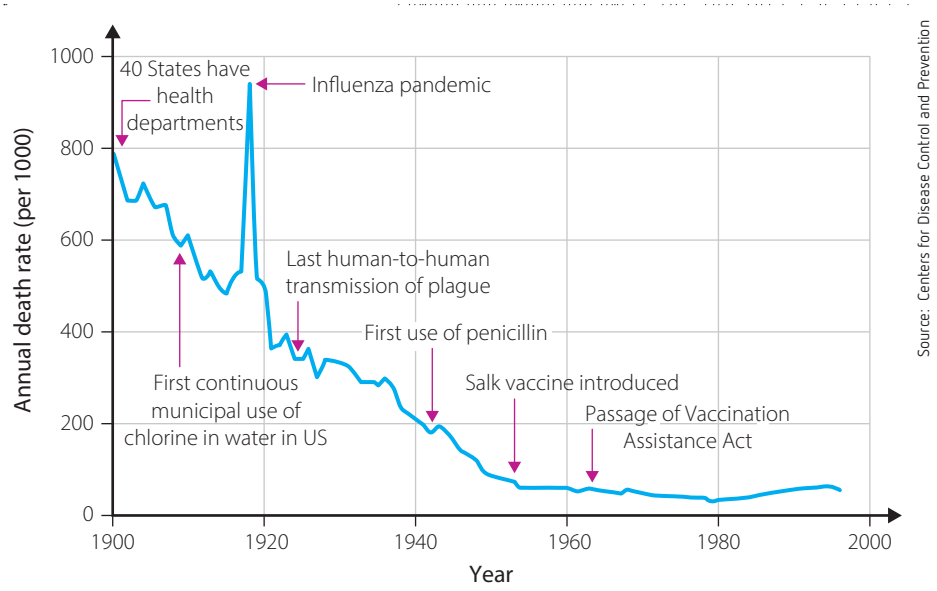

FIGURE 13.13 Death rates from infectious diseases in the USA, 1900–1996

CONCLUSION

Comment on the relationship between the introduction of penicillin and death rates from infectious disease.

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Antibiotic resistance

Bacterial resistance limits the effectiveness of antibiotics in controlling outbreaks of infectious diseases. This problem arises as an antibiotic becomes less effective over time in treating a particular bacterial disease. The rise of MRSA (methicillin-resistant *Staphylococcus aureus* or 'golden staph') in hospitals is of particular concern.

You are aware of the effect of a selection pressure on a population of organisms. In the presence of a mutation or gene (genotype) that confers resistance to an antimicrobial substance, bacteria are able to survive or grow in higher antimicrobial concentrations than most other bacterial strains of the same species (phenotype). Overuse of antibiotics during the 20th century has caused this serious problem.

KEY CONCEPTS

- Pharmaceuticals are used to treat infectious diseases caused by pathogens.
- Pharmaceuticals reduce the available pool of pathogens in hosts and therefore limit transmission rates.
- Antibiotics target bacterial pathogens by slowing their multiplication rate or killing them directly.
- Antivirals are a relatively new class of pharmaceuticals and are used in the treatment of viral diseases such as influenza and HIV/AIDS.
- Bacterial resistance to antibiotics is a major challenge and is mostly due to the overuse of antibiotics during the 20th century.

CHECK YOUR UNDERSTANDING 13.3

1. Distinguish between antivirals and antibiotics.
2. List the viruses most commonly targeted with antiviral drugs. Explain why.
3. Explain why it is necessary to understand the biology of viruses in order to manage them.
4. List three situations when antibiotics are most effective.
5. Explain how bacteria are developing resistance to antibiotics.
6. Imagine that you are a doctor in a suburban medical practice. It is winter and a particularly virulent strain of influenza is spreading through the community. Many school students are infected. A mother brings in her Year 6 child, who is coughing and sneezing and has a high temperature. Using a special diagnostic test kit, you diagnose influenza. The mother insists on antibiotics for her child, but there are no signs of a secondary bacterial infection at this stage. What advice do you give the parent? Justify your answer.

13.4 Environmental management and quarantine methods

When an outbreak of infectious disease occurs, it is important for scientists to carry out a full evaluation of their management strategies. Meticulous collection of data and other observations help scientists to evaluate such things as routines and procedures, the effect of medications, the training of staff, and other issues. In this way, recommendations can be made to make the process more efficient and successful the next time a similar event occurs.

Worksheet
Limiting the spread of an infectious disease–polio

Case study: Ebola virus disease 2014–2016

Ebola virus disease is a severe infectious disease caused by the *Ebola* virus. It is extremely contagious and causes rapid death. *Ebola* is a ssRNA virus with many subtypes. It is, however, easily preventable. It spreads when people have close direct contact with body fluids and mucous membranes (as well as items contaminated by these) from infected individuals, including sexual transmission.

Ebola virus first appeared in Africa in 1976. A large outbreak occurred in West Africa in 2014–2016. 'Patient zero' appears to have been a two-year-old child who died in southern Guinea in December 2013. Transmission is most likely through close human contact with wild animals who are reservoirs of the virus (for example, by consuming their meat) – reservoirs may be the body tissues and fluids of apes, antelope, fruit bats and porcupines. In fact, fruit bats are suspected to be the natural reservoir. Burial rituals that involve direct contact with the body of an affected person can contribute to transmission.

The incubation period is between 2 and 21 days. Symptoms start as fever and tiredness, headache and a sore throat. This progresses to vomiting, a rash, diarrhoea, and oozing of blood from the mucous membranes and stools. The bleeding is due to a coagulation failure resulting from liver damage (a healthy liver produces clotting factors).

Management of the disease is supportive, with intravenous or oral fluids to replace losses and maintain circulating blood volume, broad-spectrum antibiotics to prophylactically manage potential secondary bacterial infections, antipyretics for fever, and analgesia for pain.

The death rate of this outbreak (of 9216 confirmed cases) was around 50%. Cases were documented in six countries outside Africa.

Control of the epidemic

Preventing the transmission of pathogens in the clinical setting of a serious epidemic requires the use of procedures and protocols called *controls*. These include environmental and quarantine measures.

Administrative controls involved the initial organisation of the response and deciding which people were responsible for which parts of the overall management strategy, and obtaining funding.

Environmental and engineering controls included provision of facilities for barrier nursing (that is, providing care in strict infection control conditions) and work spaces, water and hygiene controls, hand hygiene and safe waste management (leak-proof bags and covered bins, ventilation control, sterilisation of patient care equipment and linen). Another important aspect was the provision and maintenance of personal protective equipment (PPE), including gown and coveralls, gloves, face shield, waterproof boots, head cover and respirator (Fig. 13.14). All health workers and visitors were trained to use this equipment correctly (Fig. 13.15).

FIGURE 13.14 Health workers wearing PPE bring food to patients kept in isolation at an Ebola treatment centre in Sierra Leone.

FIGURE 13.15 A health worker undergoing training in Darwin

Quarantine/isolation procedures were carried out by isolating patients in a single room or otherwise providing a space of at least three metres between patient beds. The same clinical staff were assigned to a single patient, as was all the medical equipment for their care (such as stethoscopes). All visits were restricted, except for parents of a child. Disposal of all sharps (needles, scalpels) in a puncture-proof container was vital to prevent needle-stick injuries. Contaminated environmental surfaces were cleaned

and disinfected with 0.5% chlorine solution as soon as possible after exposure to a patient. All surfaces were cleaned with detergent once a day and cleaners also wore PPE. When laboratory staff were using equipment that could aerosolise (disperse through the air) the virus (when centrifuging blood samples, for example), respirators were worn. Samples taken from infected humans and animals were handled only by trained staff and processed in properly equipped laboratories. Any tissues or body fluids for disposal were placed in clearly marked sealed bags for incineration. Any exposed person was isolated and monitored for 21 days after exposure, to help reduce transmission.

All Ebola survivors and their sexual partners received counselling and were advised to use condoms until two negative results were obtained for virus presence in semen. The virus may persist in recovered individuals in the testes, eyes and nervous system. It can be shed in breast milk. This may occur for up to nine months after infection. Border checkpoints were established with armed guards, to prevent movement of people in and out of the quarantine area (Fig. 13.16).

Longer-term measures include reducing wildlife-to-human transmission by ensuring that meat is cooked thoroughly. Wearing gloves is also recommended when handling animal reservoirs. Poverty is a driving factor for the hunting of these animals, both as food and for the pet trade.

Cultural beliefs and practices may prevent individuals seeking care at hospitals; instead they may consult traditional and spiritual healers. Working with community leaders could help to better manage this problem. Mistrust of governments and foreign workers may lead to the destruction of treatment units and even the murder of staff; this is a major limiting factor to controlling Ebola outbreaks that must be addressed in the future.

Reducing human-to-human transmission through good hygiene and environmental control procedures during and after infection is vital. Reducing the risk of sexual transmission through the use of safe sex and hygiene for 12 months after infection is one way of establishing good infection control in this area.

Outbreak containment measures may include prompt and safe burial of the dead (Fig. 13.17). After the outbreak has been contained, mandatory quarantine for workers returning from Ebola-affected countries should be a standard requirement.

An earlier and more robust response might have controlled the epidemic sooner. The appearance of cases in the USA and Europe made the international community respond more vigorously.


DOMINIQUE FAGET/Getty Image

FIGURE 13.16 Soldiers from the Liberian army monitor a border checkpoint as part of efforts to control the Ebola outbreak.


CARL DE SOUZA/Getty Images

FIGURE 13.17 An Ebola victim being buried a grave in the village of Kailahun, Sierra Leone.

KEY CONCEPTS

- Managing the environment during an epidemic reduces the pool of available pathogens.
- Quarantine measures are designed to reduce the possibility of transmission of pathogens from infected to non-infected hosts.
- Meticulous planning is part of epidemic and pandemic management, and includes education and training for all staff involved.
- Successful management of a pandemic requires goodwill and cooperation between the population and health workers.
- The Ebola epidemic of 2014–2016 is a good example of a successful strategy to limit the spread of a highly infectious viral disease.

Worksheet
Controlling an epidemic or pandemic

CHECK YOUR UNDERSTANDING 13.4

1. What is Ebola?
2. How is Ebola transmitted between hosts?
3. What is the length of the incubation period? What does this mean?
4. For each of the following categories, state what controls were put in place to prevent the transmission of the pathogen:
   - **a** environmental
   - **b** quarantine
   - **c** long-term.
5. Evaluate the effectiveness of the control measures put in place in the case of Ebola, in view of the data in Figure 13.18.

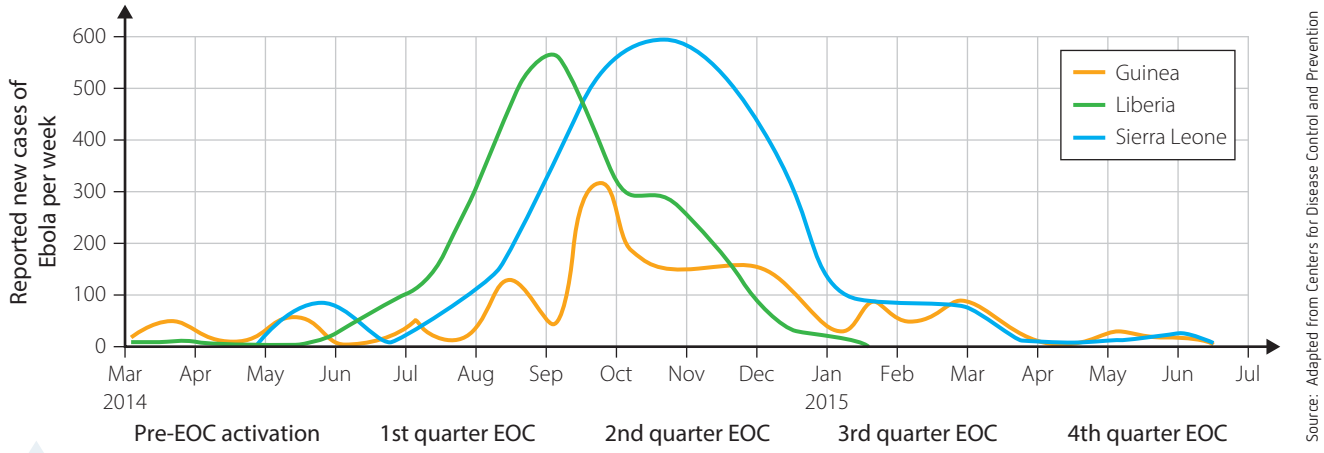

FIGURE 13.18 Reported cases of Ebola from March 2014 to July 2015 (EOC: Centers for Disease Control Emergency Operations Center)

13.5 Incidence and prevalence of a disease

The control of infectious disease outbreaks may have a new and powerful ally in the online world. When people get sick, they commonly turn to the Internet to research their symptoms. Social media is a forum for letting their friends and families know how sick they are and the details of their symptoms. Each of these interactions with the online environment is time stamped and possibly even geo-tagged (geographically identified). *Digital epidemiology* may be a way to trace, in real time, the way in which an infectious disease is moving through a population. Although this promises to be a valuable source of data, considerations such as privacy issues will prevent this happening for some time.

For now, the accumulation of real-time data on infectious diseases is currently managed in Australia by both Federal and State/Territory governments. Much progress has been made over the last 100 years, and deaths from infectious diseases have been reduced from 13% of deaths to just 1.3% in 2009. The constant threat of new and emerging diseases as well as the re-emergence of old diseases (tuberculosis, pertussis, measles) requires a robust system of data gathering and analysis.

The type of data that must be collected includes:

- incidence and prevalence of the disease – determines the pool of pathogens already present in a population
- mobility of the population – determines how easy it is for the pathogen to spread

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- percentage of the population that is immunised against an infectious disease – determines what proportion of the population are vulnerable and may act as carriers for the pathogen.

Incidence

The **incidence** of an infectious disease is the number of new cases occurring during a specified time. It can also be thought of as the infection rate, or the probability (risk) of contracting the disease. It can be expressed as a percentage (number per 100) or as a number per 100 000 (or 1000 or 10 000 and so on) of the population.

To calculate the incidence of a disease as a percentage, the following formula can be used:

$$\frac{\text{number of new cases during a specified time}}{\text{size of population at start of monitoring period}} \times 100$$

For example, of 1000 students in a school, 25 new cases of influenza appeared this week.

$$\text{incidence/risk} = \frac{25}{1000} \times 100 = 2.5\%$$

The risk of a student getting the disease that week was 2.5% or *2.5 new cases per 100 population.*

Prevalence

The **prevalence** of a disease is the proportion of the population that have the disease at a particular point in time. Whereas incidence refers only to new cases, prevalence refers to *all cases*, both previous and current. Prevalence is also expressed as a percentage.

The prevalence of a disease can be calculated using the following equation:

$$\frac{\text{all new and previous cases during a time period}}{\text{population during the time period}} \times 100$$

For example, a survey was conducted in the same school on the total number of students who had experienced influenza over the three months of winter. There were 1000 students in the school, and 150 students had contracted influenza in that winter.

$$\text{prevalence} = \frac{150}{1000} \times 100 = 15\%$$

So 15% of the student population (or 15 out of 100) had experienced the disease during winter.

Mobility

The **mobility** of a population is an important factor in the assessment of potential disease outbreaks. Humans act as carriers for pathogens and may spread the disease to new locations when they move. The *Australian Bureau of Statistics* maintains data records on the movement of people within Australia from state to state as well as migration rates from overseas. Just as importantly, social trends, such as numbers of Australians who travel overseas by plane and boat each year, are carefully monitored. In the case of Ebola, the mobility of the population played a key role in both the spread and the containment of the disease.

The **rate of immunisation** of a population is a key factor in analysing data relating to infectious disease. The Australian Government's *Immunise Australia Program* aims to promote and inform people about the value of immunisation in reducing disease transmission through a community. When a significant proportion of the population have been immunised, this creates **herd immunity**. Herd immunity relies on high numbers of individuals being vaccinated, to reduce the chances of unvaccinated individuals coming into contact with the disease-causing microbe. When a population has herd immunity, everyone in that population, including unvaccinated individuals, is protected against epidemics.

Weblink
The Australian Bureau of Statistics

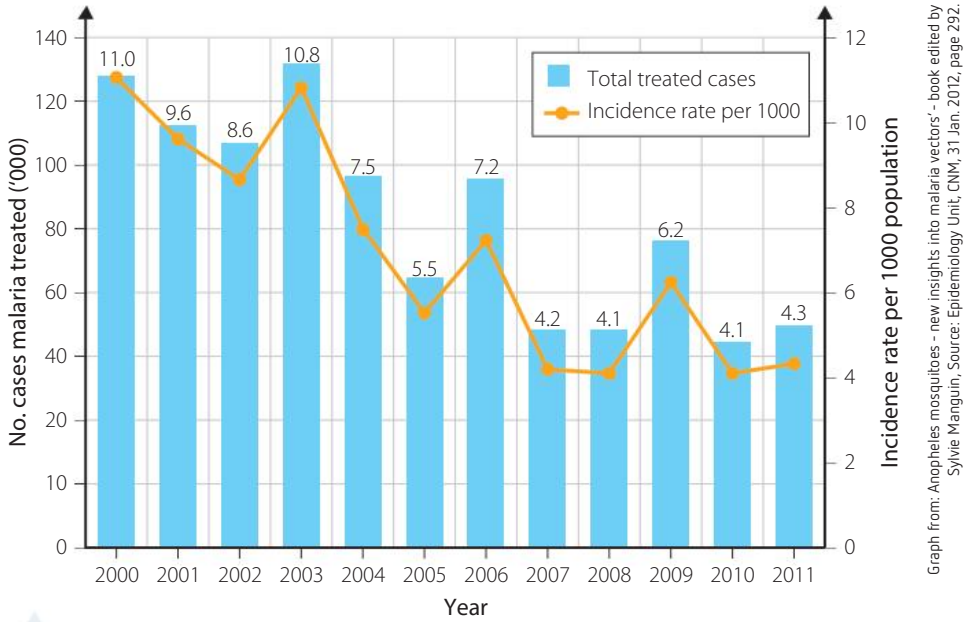
Weblink
Immunise Australia Program

INVESTIGATION 13.4

Investigating the incidence and prevalence of malaria in Cambodia

Numeracy

Malaria remains one of the leading causes of death of children in South-East Asia. The infecting microbe is *Plasmodium falciparum*, carried by various species of *Anopheles* mosquito vector. Figure 13.19 shows the total number of malaria cases treated in Cambodia between 2000 and 2011. Table 13.3 lists the Cambodian population in 2000, 2005 and 2010. Use this information to answer the questions that follow.



Graph from: Anopheles mosquitoes - new insights into malaria vectors' - book edited by Sylvie Manguin, Source: Epidemiology Unit, CNM, 31 Jan. 2012, page 292.

FIGURE 13.19 Incidence rate of malaria treated cases per 1000 population, Cambodia, 2000 to 2011

TABLE 13.3 Population of Cambodia

YEAR	POPULATION (MILLIONS)
2000	12.45
2005	13.36
2010	14.14

QUESTIONS

1 What is the general trend shown in Figure 13.19?

2 What was the incidence of malaria in the Cambodian population in:
- **a** 2000
- **b** 2005
- **c** 2010?

3 What was the prevalence of malaria in the Cambodian population in:
- **a** 2000
- **b** 2005
- **c** 2010?

4 Provide three possible reasons for the significant drop in the number of cases of malaria being treated in 2007 and 2008, from the peak in 2003.

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KEY CONCEPTS

- Data collection is vital in controlling infectious disease outbreaks.
- Epidemiological studies help to identify patterns, causes and effects of disease conditions.
- Management strategies require constant monitoring and up-to-date information.
- Important data to be collected includes the incidence and prevalence of the disease, the mobility of the population and rates of immunisation.
- Malaria and dengue fever are infectious diseases of concern in South-East Asia, and require accurate data collection if they are to be managed effectively.

CHECK YOUR UNDERSTANDING 13.5

1. Use examples to distinguish between the incidence and prevalence of a disease.
2. Out of 500 students at a school, 30 new cases of influenza appeared in a week. Calculate the incidence of the disease at this school.
3. A city has a population of 150 000. In this city, 10 000 people contracted an infectious disease. Calculate the prevalence of the disease in this population.
4. Every four years, a large number of people from nations all over the world gather at the Summer Olympic Games. Comment on the potential risks of an event like this for the potential transmission of infectious diseases. What steps might organisers take to reduce the likelihood of disease outbreaks at such events?
5. Suggest reasons why it is unwise for an individual to rely on herd immunity instead of being vaccinated against a disease.
6. What steps would you take as an individual to avoid contracting an infectious disease on a long - haul plane flight?

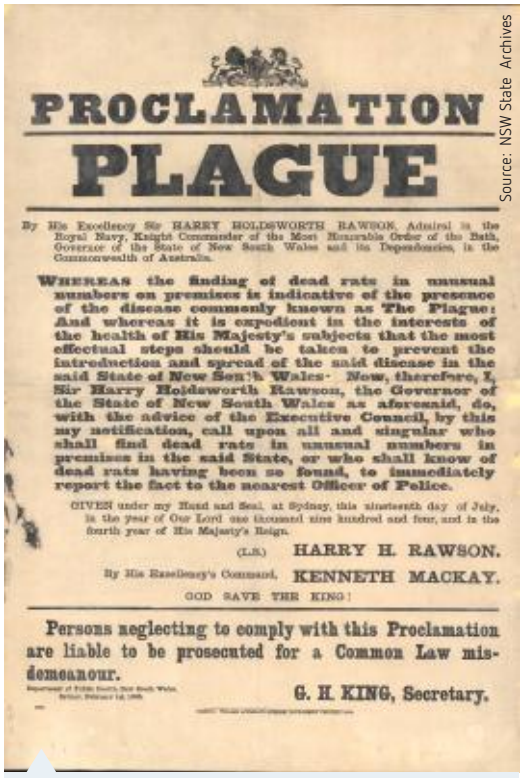
13.6 Predicting and controlling the spread of disease

The word **epidemiology** derives from the Greek words meaning 'the study of people'. In terms of infectious disease, epidemiology is the study of the incidence and distribution patterns of disease that lead to its cause, management and control.

Historic control of the spread of disease

Quarantine measures were first used in Dubrovnik, Croatia, in 1377 to control the outbreak of plague. The first permanent plague hospital, or *lazaretto*, was opened on the island of Santa Maria di Nazareth in 1423. In 1467, a similar hospital for leprosy patients was opened in Marseille, France. *Lazarettos* were strategically located so that they had a natural barrier such as a sea, river or mountain, to separate them from populated areas. Similar strategies were used during the global SARS (severe acute respiratory syndrome) outbreak in 2003, when sufferers were isolated either in hospitals or in their homes. Such strategies were used to contain infection and to delay the spread of the disease; from a social point of view, they also helped avert panic and maintain social stability.

It would probably surprise many young people to know that Sydney has experienced many epidemics during its short history: measles (1866–67), scarlet fever (1876–76), smallpox (1789, 1881–82, 1913–17), the Asiatic Flu pandemic (1890–91), plague (1900) and the Spanish influenza pandemic (1918–19). The responses to these early epidemics laid the foundations for the public health system in New South Wales.


Source: NSW State Archives

PROCLAMATION

PLAGUE

By His Excellency Sir HARRY HOLDSWORTH RAWSON, Admiral in the Royal Navy, Knight Commander of the Most Honorable Order of the Bath, Governor of the State of New South Wales and its Dependencies, in the Commonwealth of Australia.

WHEREAS the finding of dead rats in unusual numbers on premises is indicative of the presence of the disease commonly known as The Plague: And whereas it is expedient in the interests of the health of His Majesty's subjects that the most effectual steps should be taken to prevent the introduction and spread of the said disease in the said State of New South Wales: Now, therefore, I, Sir Harry Holdsworth Rawson, the Governor of the State of New South Wales as aforesaid, do, with the advice of the Executive Council, by this my notification, call upon all and singular who shall find dead rats in unusual numbers in premises in the said State, or who shall know of dead rats having been so found, to immediately report the fact to the nearest Officer of Police.

GIVEN under my Hand and Seal, at Sydney, this nineteenth day of July, in the year of Our Lord one thousand nine hundred and four, and in the fourth year of His Majesty's Reign.

(L.S.) HARRY H. RAWSON.

By His Excellency's Command, KENNETH MACKAY.

GOD SAVE THE KING!

Persons neglecting to comply with this Proclamation are liable to be prosecuted for a Common Law misdemeanour.

G. H. KING, Secretary.

FIGURE 13.20 Plague Proclamation poster displayed in Sydney, 1 February 1905

INVESTIGATION 13.5

Historic control of a disease outbreak

Information and communication technology capability

Literacy

Worksheet
Preventing the spread of an infectious disease - polio

John Snow is known as the 'father of field epidemiology'. He proposed and tested hypotheses regarding the 1854 epidemic of cholera in London (Golden Square). His use of maps to record deaths was a key factor in pinpointing the source of the pathogen to a single water pump in Broad Street.

AIM

To evaluate the contribution of the work of John Snow to the prediction and control of infectious disease spread

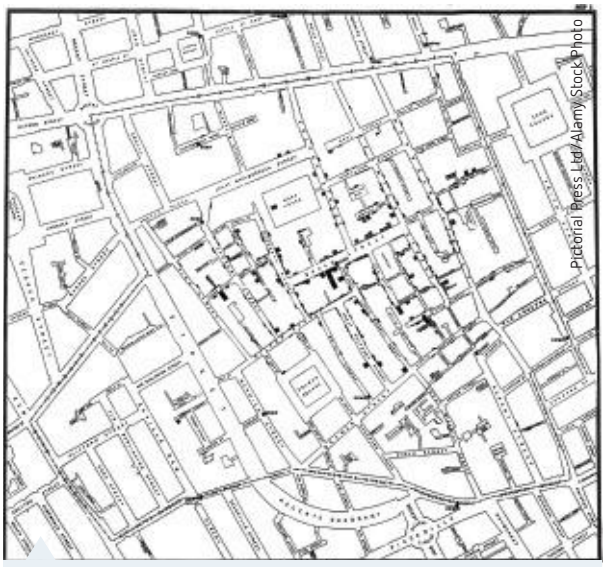

Pictorial Press Ltd/Alamy Stock Photo

FIGURE 13.21 John Snow's map of cholera cases in London

METHOD

1 a Use printed and online sources to investigate the following aspects of John Snow's identification of the cause of cholera:
   - i the date of the outbreak
   - ii the location
   - iii symptoms observed in patients
   - iv statistics on the prevalence of the disease during this outbreak
   - v historical/cultural explanations for the disease at the time and effectiveness of control methods
   - vi Snow's methodology in approaching the study of the disease
   - vii statistical data including maps and tables
   - viii Snow's proposed cause and solution, and responses by the authorities at the time.

   b Evaluate the effectiveness of John Snow's epidemiological approach to the Golden Square cholera outbreak.

2 Record all data in the appropriate format.

3 Present a reference list in the appropriate format.

4 Present your findings in a format agreed upon with your teacher.

DISCUSSION

1 Do you agree that John Snow is the 'father of field epidemiology'? Use what you have learned about him to justify this statement.

2 Evaluate John Snow's approach to disease control against the scientific method.

3 What can modern-day epidemiologists learn from John Snow?

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Cultural control of the spread of disease

Culture refers to integrated patterns of human behaviour, including the language, thoughts, communications, actions, customs, beliefs, values and institutions of racial, ethnic, religious or social groups. Some cultural beliefs can hamper attempts to prevent and contain a disease.

In 2014, the Ebola outbreak in Africa was able to spread because of a combination of poverty, famine and corruption, along with the traditional ritual of paying respects to the deceased through close contact with the corpse. Control measures needed to include educating the public to abandon these traditional cultural belief systems.

Different cultures have developed ways of controlling disease, even without fully understanding the true nature of it. In the Philippines, traditional foods play a large part in preventing and curing disease. Garlic and onions are believed to lower blood pressure. It is now known that onions contain quercetin, which is a natural supplement that aids in lowering blood pressure. Mosquito nets were said to have been used by the Egyptian queen Cleopatra (69–30 BCE) even though the connection between malaria and mosquitoes was not known.

Intercultural understanding

Worksheet
Preventing mosquito-borne diseases by using vector control

Epidemiology is discussed in detail in Chapter 16.

KEY CONCEPTS

- Historically, different strategies including quarantine have been used to prevent the spread of disease.
- John Snow is known as the 'father of field epidemiology'.
- Disease control is influenced by culture, and this needs to be taken into account when developing control measures.

Weblink
Sociocultural factors in the control and prevention of parasitic diseases

CHECK YOUR UNDERSTANDING 13.6

1. Define 'epidemiology'.
2. How does quarantine assist in controlling infectious diseases?
3. Who is John Snow and why is his work with cholera considered important?
4. How does culture influence the spread and treatment of infectious diseases?
5. Use the weblinks to find out what sociocultural factors influence the spread and treatment of infectious diseases.

13.7 Aboriginal protocols in the development of medicines

Many indigenous cultures around the world have used traditional methods to treat infectious and non-infectious diseases. Aboriginal Australians are the custodians of a rich and detailed knowledge base of medicinal native Australian herbs, fruits and vegetables. Traditionally, Aboriginal people lived healthy lives. However, on occasion there was a need to manage wounds from burns, stings and bites as well as food poisoning from meats.

Substances from plants such as tannins, mucilage, oils, latex and alkaloids were used for medicine. The plant material was usually crushed and used as a **poultice** or infused with water to drink. Animal fat was often incorporated into the plant material. This increased the fat solubility of the plant substance and increased absorption rates into the tissues.

There has been a renewed interest in traditional Aboriginal medical knowledge. Databases are being compiled by different groups to ensure that these traditional approaches to disease control are not lost. One example of this is the Macquarie University Indigenous Bioresources Research group – see the weblink.

Weblink
Social and cultural factors in the successful control of TB

Weblink
Customary Medicinal Knowledgebase (CMKB) – database of native plants

Scientists are now discovering that many **bioactive compounds** are contained in these traditional **bush medicines**. Some contain antimicrobial properties that are useful in managing certain infectious diseases. Alkaloid compounds from the Moreton Bay Chestnut (*Castanospermum austral*) or black bean are showing promise in the management of HIV/AIDS. It must be remembered that many of these plants contain potentially deadly compounds as well. Aboriginal people use specialised preparation techniques to minimise harmful effects.

FIGURE 13.22 Smokebush, used as a medicinal plant by Indigenous peoples

In 1798, English botanist James Smith named the genus *Conospermum*, meaning 'cone seed'. These plants are commonly known as smokebush and grow mostly in south-west Western Australia, with some species occurring in NSW and Tasmania. Some species of this plant have very big and woolly white flowers that resemble drifting smoke (Fig. 13.22). These plants are a member of the *Proteaceae* family. Indigenous people have used the smokebush for healing. Scientists have now investigated the properties of this plant and its potential uses against cancer and HIV/AIDS.

Bush medicine is a new branch of horticulture that promises to be a fertile area for the development of new and effective treatments against a range of pathogens. Scientists must separate the useful from the deadly before these treatments can be used commercially.

INVESTIGATION 13.6

Aboriginal protocols in medicines and biological materials

Aboriginal and Torres Strait Islander histories and culture

Sustainability

Ethical understanding

AIM

1 To gather and process data on the current use of common biological materials by Aboriginal people (bush medicine)

2 To establish how Indigenous cultural and intellectual property is to be protected

METHOD

1 Use online and printed resources to gather information on the properties and uses of the plants and animals listed in the table below. You are not limited to these – your teacher will guide you as to the number of materials to be researched.

2 Be sure to use publications that are authored or endorsed by Aboriginal or Torres Strait Islander people. You may be able to involve local Aboriginal communities when gathering information. It is *traditional* uses that you are investigating in this activity.

3 Use a table like the one shown here to record your results.

PLANT OR ANIMAL	TRADITIONAL PROPERTIES AND USES AS ANTIMICROBIALS	PHOTO
Emu bush leaves		
Tea tree leaves		
Kakadu plum fruit		
Witchetty (*witjuti*) grub		

»

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RESULTS

You could present your findings in a table like the one on the previous page.

QUESTIONS

1 Why is it preferable to gather information from sources that are authored by Aboriginal people? Outline some of the possible problems when using sources that are not from the Aboriginal community.

2 Suggest reasons why it is more difficult to find information on bush medicine than Western forms of medicine.

3 Discuss the role of traditional medicine in the modern world. Is there value in retaining the knowledge that has been passed down for thousands of years?

4 Much of the knowledge about traditional medicines is handed down orally and has not been written down. Suggest a way in which we might be able to record this knowledge for future generations so it is not lost forever.

5 What does the law say about indigenous people, biodiversity and their **intellectual property**? (See the weblink.)

Use the Internet to answer the following questions.

6 To take out a **patent** on a piece of technology or idea, what steps must be taken? What features of intellectual property must be satisfied for a patent to be awarded?

7 Do you consider Western intellectual property laws to be biased against the rights of indigenous peoples? Use evidence from your research to justify your answer.

8 Research the re-discovery by Western scientists of the WA smokebush in the 1960s. What financial and legal arrangements were made between the WA government and the Australian pharmaceutical company AMRAD? Did Aboriginal people receive any recognition of their traditional knowledge or financial benefit from the exploitation of the plant?

Weblink
Indigenous people and intellectual property

CONCLUSION

Summarise the ethics using bush medicines, and their value to the health and wellbeing of Aboriginal people.

KEY CONCEPTS

- Traditional medicine, such as Aboriginal bush medicine, is a valuable source of knowledge that needs to be preserved for future generations.
- The intellectual property rights of Indigenous peoples are not well supported in Australia as yet.

CHECK YOUR UNDERSTANDING 13.7

1 An overseas tourist finds a previously unknown species of plant in the outback. They claim that chewing the plant relieved their indigestion. The plant grows in an area traditionally owned by Aboriginal peoples.

a Who owns the plant? What does the law say?

b Does the tourist have any intellectual property rights to this biological material? Could they take out a patent and sell the plant as an indigestion medicine?

13 CHAPTER SUMMARY

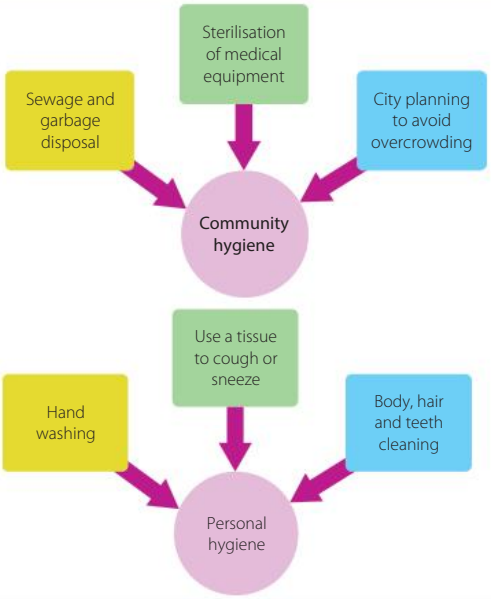
Prevention, treatment and control of disease: How can the spread of infectious diseases be controlled?

Monitoring and control

The three levels of disease monitoring and control

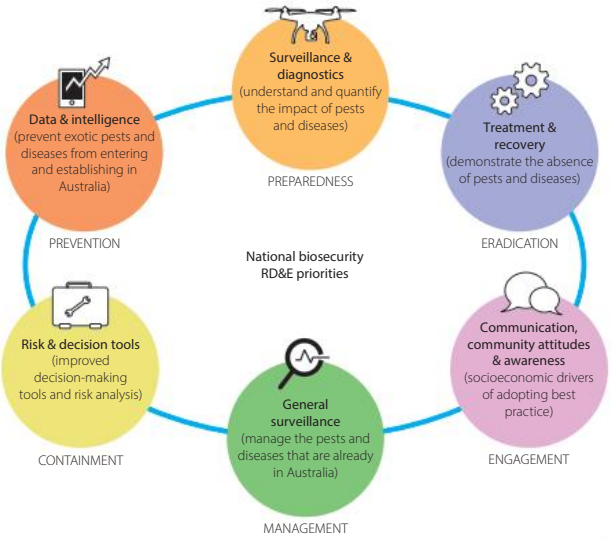
Factors affecting disease outbreaks

Personal and community hygiene



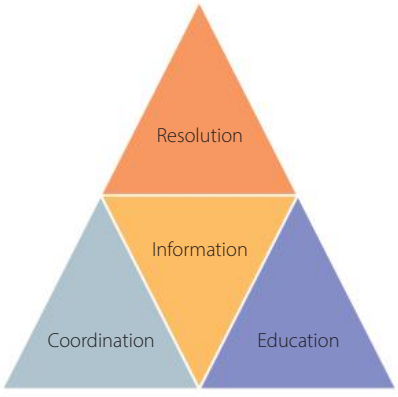
Biosecurity

DAWR has many roles in keeping infectious diseases out of Australia



Public health campaigns

Three levels of approach to infectious disease limitation



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Antimicrobial drugs

CLASS OF MEDICATION	PATHOGENS TARGETED	EXAMPLES
Antibiotics (antibacterials)	Bacteria	Penicillins, tetracyclines, polymixins, sulfonamides, cephalosporins
Antivirals/ antiretrovirals	Viruses	• Tamiflu® (oseltamivir) used to treat influenza (antiviral) • Abacavir (antiretroviral used in HIV/AIDS treatment)
Antifungals	Fungi	Fluconazole, amphotericin B, caspofungin
Antiprotozoals	Protozoa	Doxycycline, metronidazole, mefloquine

Protocols

Control of an epidemic (e.g. Ebola)

Administrative protocols	**Environmental and engineering protocols**	**Quarantine/isolation protocols**
Who is involved and and how will it be funded	Barrier nursing/hygiene	Infected people and contaminated objects do not leave the site (short and long term)

Incidence and prevalence

Incidence

The *incidence* of an infectious disease is the number of new cases occurring during a specified time.

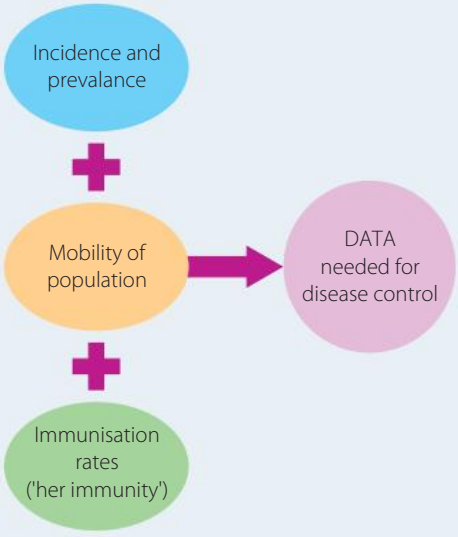
To calculate the incidence of a disease can as a percentage, use the following formula:

$$\frac{\text{number of new case during a specified time}}{\text{population at start of monitoring period}} \times 100$$

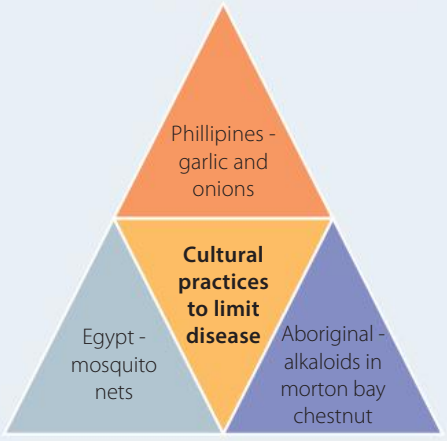
Prevalence

The *prevalence* of a disease is the proportion of the population that have the disease at a particular point in time.

$$\frac{\text{all new and previous cases during a time period}}{\text{population during the time period}} \times 100$$



Cultural controls



13 CHAPTER REVIEW QUESTIONS

Review quiz

1 Explain why limiting infectious disease transmission does not rely on one factor alone but a combination of factors.

2 Assess the effect of increased mobility of populations in the transmission of infectious diseases.

3 Imagine a scenario where garbage is not collected from homes and businesses for one month. How could individual householders manage their own disease risk during this crisis?

4 Justify research into alternatives to antibiotics in the treatment of bacterial diseases. Is it possible to avoid using pharmaceuticals entirely?

5 Compare the disease transmission risk between an overcrowded city such as Jakarta with a spacious city such as Perth, WA. Assess the factors that are most likely to be of greatest concern in an overcrowded city.

6 Compare and contrast tissues and handkerchiefs in terms of their ability to limit transmission of infectious diseases.

7 Evaluate the effectiveness of a current public health campaign in limiting disease transmission. What aspects would you change to increase awareness and compliance?

8 Assess Australia's role in allowing Indigenous people to claim intellectual property rights on biological materials.

9 Airborne transmission of pathogens such as pulmonary tuberculosis, measles and chickenpox is of great concern to public health authorities. Assess the reasons why airborne pathogens require stricter hygiene precautions in hospitals than waterborne diseases such as giardia.

10 How would you advise a worker who is responsible for the safe disposal of biological waste in a hospital? What types of protocols would protect the worker and the public from this potential source of pathogens?

11 Australia is relatively free of natural disasters such as earthquakes and hurricanes, where infectious diseases often increase. What role do you think Australia should play in offering assistance to other nations that experience natural disasters? Do we have a 'duty of care' to other countries?

12 If pathogens are the cause of infectious disease, explain why scientists allocate resources to investigating the features of target host organisms. What potential benefit could arise from understanding the host's involvement in disease spread?

13 In 2015, a famous Hollywood couple brought their Yorkshire terriers into Australia on a private plane, bypassing quarantine. The then Australian Deputy Prime Minister and Minister for Agriculture was adamant that the dogs, Pistol and Boo, needed to be immediately removed from the country or face being euthanised. The couple eventually removed the dogs and issued an apology in the face of legal action by the Australian Government.

Weblink
Johnny Depp's dogs

In your opinion, did the Australian Government overreact? Assess the claim made by the minister that the dogs posed a biosecurity risk. Read more about it in the weblink.

14 Fears are spreading about the appearance of a new 'superbug' strain of the malarial parasite *Plasmodium falciparum*. The new strain has been reported in five South-East Asian countries but is of particular concern along the border between Cambodia and Thailand. Why is the likelihood of the spread of this pathogen greater now than in past centuries? Assess some of the factors involved.

15 In 2017, a new study questioned the long-held belief of scientists that it is essential to complete the entire course of antibiotics you are prescribed. The opinion piece was published in the *British Medical Journal*. Would you take this report at face value? What more information would you like to know? Where would you look for further information on this very important topic?

16 Antibiotics have been found in samples taken from Australian waterways. Suggest some of the ways in which antibiotics used in animals and humans could make their way into our waterways. What are the possible consequences of this for the health of the environment?

17 Describe what the world would be like if antibiotics and antivirals did not exist.

18 In what way do antibiotics and antivirals affect evolution by natural selection in humans and animals?

19 Pharmaceutical companies are often criticised for the high price of pharmaceuticals. Use secondary resources to find out some of the stages involved in the development of a new chemical entity. Find out how much it costs in time and money, and then assess whether the high prices are justified.

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20 Imagine that you are transported back to the time of Hippocrates (400 BCE). Assuming you can speak ancient Greek, what advice would you give Hippocrates about investigating the causes of a sudden outbreak of an infectious disease in the local community?

21 During the Spanish Flu outbreak of 1918–1919 in Sydney, most workplaces and all schools were closed. Describe what advantages we have today, if the same outbreak were to occur now. Why could most work and schooling continue relatively unaffected?

22 When cultural practices clash with accepted hygiene practices, how should the situation be resolved? Brainstorm some ideas about ways in which both science and culture can work out such issues. Suggest an example of a situation where this occurs.

23 Imagine that the rates of a certain type of waterborne disease are higher in a particular area in a big city in South-East Asia.

- **a** Outline the immediate steps you would take to investigate the cause.
- **b** Make a list of the important data you would collect.
- **c** What recommendations would you make regarding limiting transmission of the disease?

24 You are a doctor and you are interviewing a patient who has been using traditional bush medicine to manage their headaches. The patient reports that 50% of the time the headache disappears. Outline an approach you would take to investigate the efficacy of the traditional medicine. What information do you need to collect to be able to assess whether the bush medicine is actually having an effect?

Exam preparation

MODULE 7: INFECTIOUS DISEASE

Answer the following questions.

1 A patient presents to their doctor with a red, raised and itchy patch of skin that is weeping fluid. The doctor suspects an infection but is not able to pinpoint which pathogen is responsible. The doctor presses a microscope slide onto the wound (an impression smear) and places the slide under a microscope for examination.

- **a** Which classes of pathogens would be visible under the light microscope?
- **b** What steps need to be taken to make the tissue more easily seen on the slide?
- **c** What types of cells might the doctor see on the slide if the body has launched an inflammatory response against the pathogen?

2 You have been asked to design a new state-of-the art animal shelter for your community.

- **a** What principles and protocols would you include in your design to reduce the likelihood of infectious disease transmission between the animals in the shelter?
- **b** You notice that some of the dogs in a pen at the end of a run are sneezing and coughing. Outline the immediate steps you would take. Justify your actions in terms of your understanding of the principles of disease transmission.
- **c** What advice would you give those who work in animal shelters regarding hygiene practices to limit the transmission of infectious diseases?

3 Many people in Sydney work in offices where large numbers of staff are crowded into small spaces. Assess this design in terms of disease transmission. As an office manager, re-design an office space to reduce the likelihood of colds and flu getting out of hand in winter. Why might it make financial sense to a company to spend money on a refit of their office space for this purpose?

4 Another student who does not study Biology argues that plants get sick because they do not have an immune system. How would you persuade them that this is an inaccurate assessment? Use specific examples.

5 Pharmaceuticals such as corticosteroids are useful in the suppression of the inflammatory response in certain conditions (e.g. autoimmune disease). Patients are administered an initial dose, which is gradually reduced over time. Justify the practice of doctors prescribing the minimum dose of a corticosteroid that suppresses the autoimmune disease.

6 'It is possible that immune responses that are meant to protect us sometimes end up harming us.' Justify this statement, using examples you have studied.

7 Despite advances in technology and education programs, many sexually transmitted infectious diseases are on the rise again. Assess the reasons for this. Justify the statement that infectious disease control involves not just good medicine but also compliance with guidelines.

8 Evaluate the role of government agencies in control of infectious diseases.

9 Assess the role of social media as a tool for the collection of data on infectious disease rates in populations.

10 Discuss the role of indigenous populations around the world in contributing to the management of infectious diseases.

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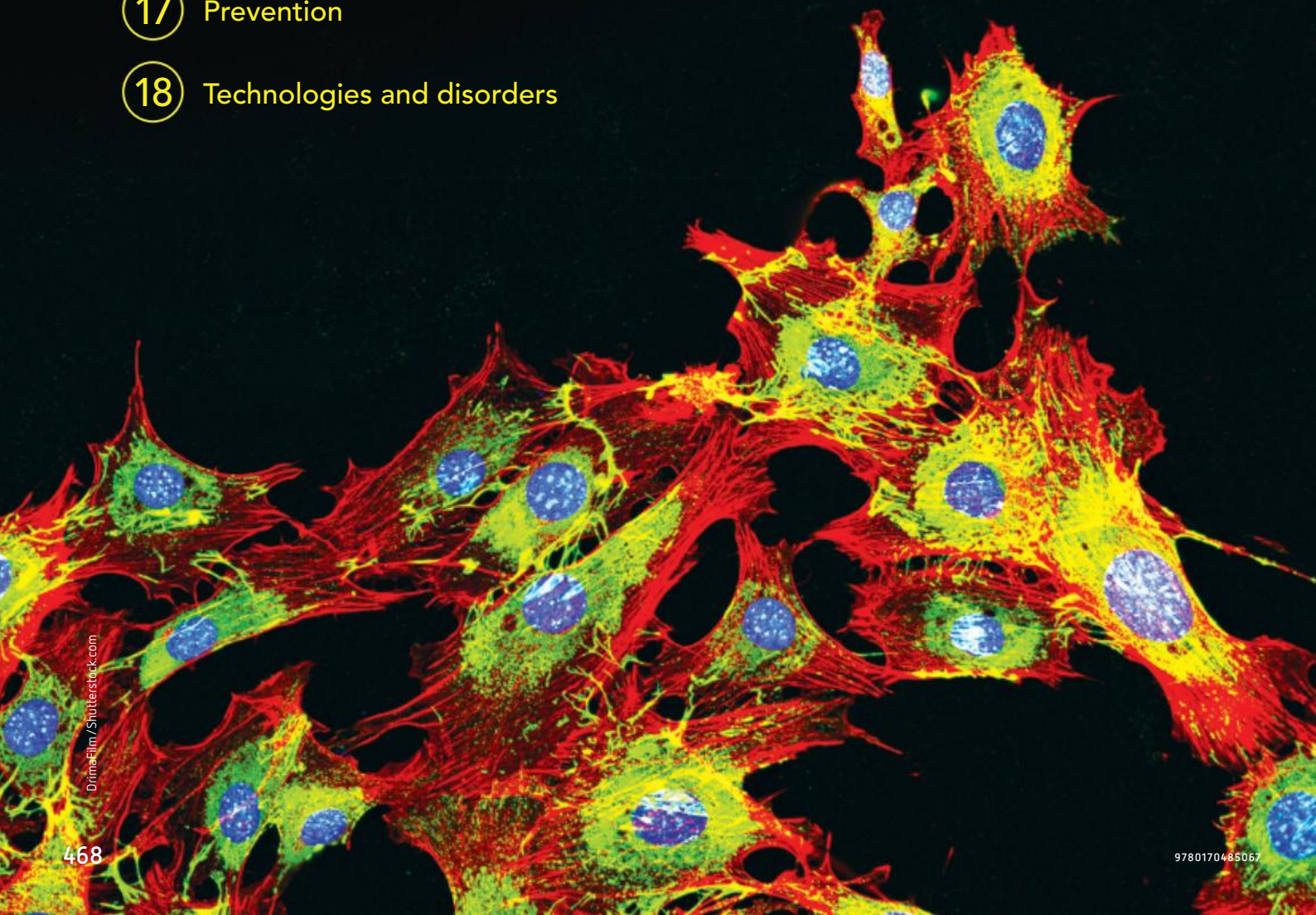
DEPTH STUDY SUGGESTIONS

- Microbial analysis of drinking water. Is bottled water free of microbial contamination?
- Locate a tree doctor in your area. What are some of the latest techniques in treating infectious disease and repairing wounds in plants?
- What is the effect of microgravity on the immune responses in plants? Send a plant experiment to the International Space Station.
- If you have a vet school nearby, make contact with them and ask to be involved in some aspect of research into the immune response in companion animals – for example, do birds respond in the same way as dogs and cats to pathogens? What about fish?
- New discoveries in the use of stem cells for autologous transplants – can we store stem cells when we are young and use them in the future in case of accident or illness?
- Vaccine production for new and emerging diseases.
- Is the innate immune system *really* incapable of forming memory? What are the latest animal models telling us?
- Does sneezing into your elbow really reduce the number of pathogens you release into the air?
- How do vets manage infectious disease transmission in their hospitals? Contact your local veterinary surgeon and ask to visit. Design your own veterinary hospital to ensure minimal risk of transmission of pathogens. What design principles are important in waiting rooms, consultation rooms and surgical suites? How are patients with highly infectious diseases (such as parvovirus-infected dogs) managed?
- How are infectious diseases controlled in refugee populations in camps? Design a refugee camp to minimise the transmission of infectious diseases.
- Is simple hand washing enough to eliminate the spread of influenza through a school population?
- Assess the effectiveness of hospital hygiene protocols. Visit your local hospital and learn the principles of design. Choose a particular aspect and redesign it to make it cheaper and more convenient, to increase compliance by doctors, nurses and visitors, or some other aspect, e.g. hand washing stations.
- What potential benefits could come from using virtual reality / 3D worlds in teaching school and university students the principles of infection control? Is it possible to design an 'outbreak scenario' in the virtual world?
- Is a doctor's surgery waiting room a place where infectious diseases are transmitted? Visit your local doctor and inquire about the risks and protocols undertaken to reduce the transmission of pathogens in the waiting room. Formulate some suggestions of your own to rethink the waiting room.
- Visit your local childcare centre and learn about the protocols and designs to reduce disease transmission between children. How would you design your own childcare centre to improve these procedures? Think about the materials and the elements of design to make the centre easy to keep clean and yet still safe and appealing for the clients.

» MODULE EIGHT

NON-INFECTIOUS DISEASE AND DISORDERS



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14 Homeostasis

INQUIRY QUESTION

How is an organism's internal environment maintained in response to a changing external environment?

Students:

- construct and interpret negative feedback loops that show homeostasis by using a range of sources, including but not limited to: (ACSBL101, ACSBL110, ACSBL111) CCT ICT
  - temperature (ACSBL098)
  - glucose
- investigate the various mechanisms used by organisms to maintain their internal environment within tolerance limits, including:
  - trends and patterns in behavioural, structural and physiological adaptations in endotherms that assist in maintaining homeostasis (ACSBL099, ACSBL114) ICT
  - internal coordination systems that allow homeostasis to be maintained, including hormones and neural pathways (ACSBL112, ACSBL113, ACSBL114)
  - mechanisms in plants that allow water balance to be maintained (ACSBL115) ICT

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
- **14.1** A secondary source investigation into the regulation of glucose concentration in the blood
- **14.2** A secondary-source investigation to graph action potential
- **14.3** A first-hand and secondary-source investigation to investigate the structure of neurons, and to observe and model neurons
- **14.4** A first-hand and secondary-source investigation into mechanisms in plants to maintain water balance

Worksheets
- Brain dissection
- Hormonal control of homeostasis
- Adaptations to endothermy
- Mechanisms to conserve water in plants

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Optimal metabolic efficiency in an organism is essential for the overall health and wellbeing of that organism. The maintenance of a relatively constant internal environment in an organism, called *homeostasis*, is essential so that enzymes can function effectively, which in turn optimises metabolic efficiency.

Regulatory systems in plants and animals act to maintain balance in their internal environments. Some of the conditions that these systems regulate are: blood pressure, temperature, pH, water, concentrations of salt, glucose, oxygen and carbon dioxide, and metabolic wastes.

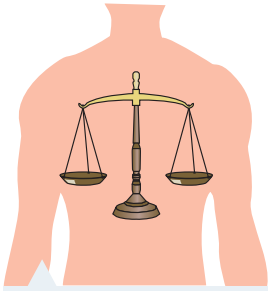

FIGURE 14.1 Maintaining a constant internal environment is important for the maintenance of health and wellbeing.

14.1 Homeostasis

Homeostasis is the maintenance by an organism of a relatively constant internal state, regardless of external changes in the environment. The word *homeostasis* comes from the Greek words *homoios*, meaning 'like' or 'the same' and *stasis*, meaning 'state'. This implies a state of balance or constancy, where conditions 'stay the same' in the internal environment of living organisms, to allow them to function at their optimal metabolic efficiency despite changes in the external environment.

The importance of homeostasis

Living organisms are made of cells, which must function efficiently to maintain life. In order to bring about optimal metabolic efficiency, all chemical reactions within cells must occur efficiently and be effectively coordinated. These reactions are catalysed by enzymes, which are very sensitive to changes in their environment. It is essential therefore that internal conditions be maintained at a level that allows the optimal functioning of enzymes to ensure that optimal metabolic efficiency is maintained.

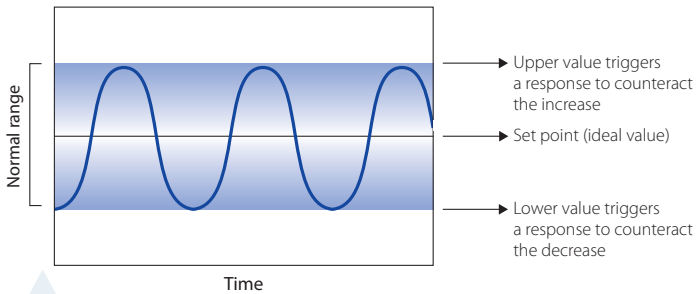

FIGURE 14.2 Graph showing homeostasis as the maintenance of a relatively constant internal environment. The value of the variable fluctuates within a narrow range of tolerance limits around a set point.

Enzymes are extremely sensitive to the temperature and pH of the internal environment, and changes to these as well as to the concentration of substrates in the reactions will affect their activity. It is also very important to maintain water and salt concentration, along with the removal of wastes such as carbon dioxide and other metabolic wastes, in order to maintain internal conditions that allow maximum enzyme activity.

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Maintenance of homeostasis

Homeostasis involves an enormous amount of coordination and control in an organism. In mammals, both the nervous system and the endocrine (hormonal) system are involved in homeostasis. The phrase 'relatively constant internal state' used in the definition of homeostasis indicates that some change in the internal environment is allowed for. Variables in the internal environment, such as temperature or glucose concentration, are maintained within a narrow range, known as the **tolerance limits**. Each of these variables has an ideal or normal value, called the **set point**.

Homeostasis is maintained as long as there is only a narrow range of fluctuation within the tolerance limits around the set point. If the fluctuation is larger and exceeds the upper or lower tolerance limits, a mechanism comes into operation to return the body to the normal range.

KEY CONCEPTS

- Optimal metabolic efficiency is essential for an organism's overall health and wellbeing.
- The maintenance of a relatively constant internal environment in an organism is essential so that enzymes can function effectively, which in turn optimises metabolic efficiency.
- Homeostasis is defined as the maintenance by an organism of a relatively constant internal state, regardless of external changes in the environment.
- Both the nervous and endocrine systems are involved in coordinating and controlling the process of homeostasis.
- All internal conditions have an ideal or set point. The internal conditions are maintained within narrow limits (tolerance limits) around this set point.

The negative feedback system

Homeostasis is brought about in two main stages:

1. *Detecting change*: sensory cells or **receptors** within the body detect a change in a particular component of the internal environment, such as the temperature of the body or the pH of the blood. This change in the internal environment is called a **stimulus**.
2. *Counteracting the change*: a **response** occurs that will reverse (or counteract) the change. This response is brought about by the **effector** organs (such as muscles or glands) and will restore the body to its relatively constant internal state.

The link between these two stages is the **control centre**. The control centre, which is responsible for maintaining fluctuations around the set point, receives information from the receptors about a change in a condition either too far above or below the set point. It then determines an appropriate response and sends a message to the effectors to carry out activities that will bring about a response that counteracts (or reverses) the stimulus and returns the levels to the set point. Once acceptable levels are achieved, as detected by the receptors, this information is fed back via the receptors to the control centre, which then directs the effectors to cease their actions.

This self-regulating system is known as a **negative feedback mechanism**. The message from the receptors to the control centre and the response that is directed by the control centre to change the original stimulus in some way is the feedback. This feedback is called 'negative' because the response counteracts the stimulus – it corrects the change and returns it to the set point. This process can be represented by a generalised negative feedback loop, as shown in Figure 14.3.

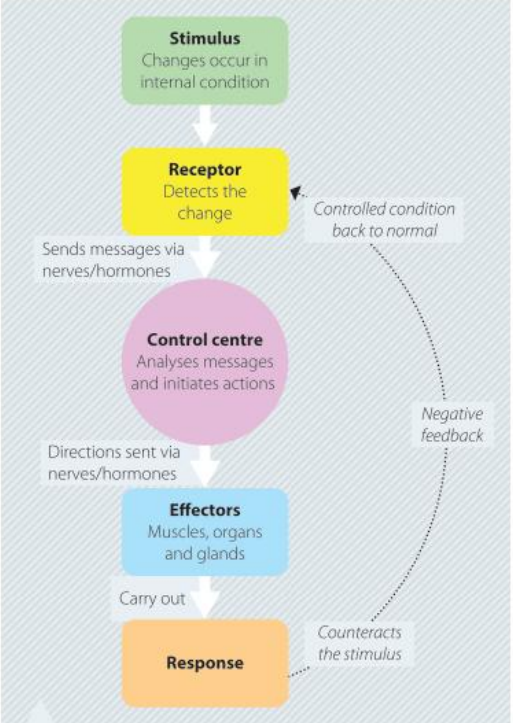

FIGURE 14.3 Generalised negative feedback loop showing the process by which the body maintains homeostasis

Coordination of the homeostatic mechanism

The negative feedback systems that maintain homeostasis can be coordinated by the nervous system, the endocrine system or a combination of both, depending on which particular condition in the internal environment is involved.

The nervous system communicates within the body by relaying information throughout the body in the form of nervous impulses. The endocrine system does this by the release of chemical messengers called hormones.

You will learn more about the nervous and endocrine systems and the hypothalamus on pages 487–493.

The **hypothalamus**, a region in the lower central part of the brain, is an important control centre for maintaining homeostasis. It contains receptors for certain factors such as body temperature, acts as a link between the nervous and endocrine systems, and sends messages to the effectors to carry out responses necessary for the maintenance of many homeostatic conditions.

Weblink
Negative feedback loops
Watch the animation and outline the role of the different components of negative feedback loops.

KEY CONCEPTS

- The two stages by which homeostasis occurs are:
  1 detecting the change
  2 counteracting the change.
- The control centre receives information from the receptors, analyses it and sends instructions to the effectors to carry out a response that counteracts (reduces) the stimulus.
- Once the receptors detect acceptable levels, the control centre directs the effectors to cease their action.
- This process is called a negative feedback mechanism.
- The hypothalamus is an important control centre in a negative feedback system.
- Messages are carried as nerve impulses or as hormones. Both the nervous and endocrine systems are involved in the coordination of a negative feedback system.

Negative feedback loops

Specific negative feedback loops can be used to show the way in which the body maintains set levels for specific internal conditions. For example, the regulation of body temperature can be summarised by a negative feedback loop, as shown in Figure 14.4.

In the negative feedback mechanism indicated by this loop, **thermoreceptors** detect changes in temperature (stimulus). Some of these receptors are in the skin and others are in the hypothalamus, where they monitor the temperature of the blood as it circulates through the brain. The receptors in the hypothalamus are sensitive to extremely small temperature changes.

In this case, the hypothalamus is also the control centre for temperature regulation in the mammalian body, which means that the receptors do not have to transmit the information very far in order for a response to be initiated.

Cooling the body

When receptors detect an increase in the body temperature (stimulus), messages are sent to the **anterior** (front) area of the hypothalamus, which is the heat-loss control centre. Messages are then sent via the nervous system to effectors that initiate processes to lose heat and cool the body down.

These processes include:

- **vasodilation**, which involves the blood vessels dilating (expanding), bringing blood closer to the skin and allowing heat to escape (Fig. 14.5)
- activation of the sweat glands to secrete liquid sweat, which removes heat from the body when it evaporates (changes state from liquid to gas) (Fig. 14.6)
- activation of the thyroid gland to lower the rate of metabolism in cells by reducing the amount of the hormone thyroxin produced. This generates less heat in the body.

  The combined effect of these processes is a decrease in body temperature, which counteracts the stimulus and returns the temperature to its set point (or normal level).

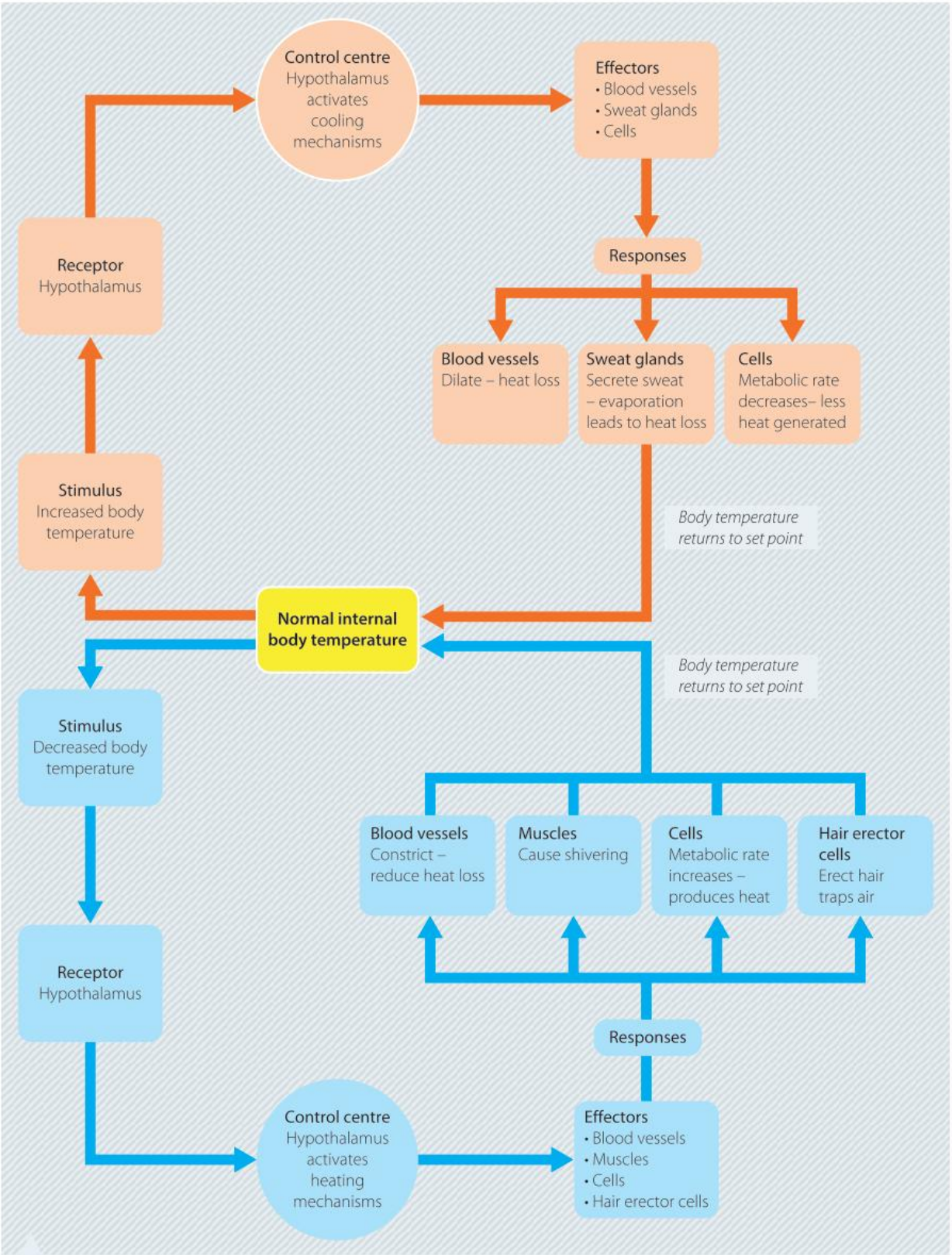

FIGURE 14.4 Negative feedback loop, showing internal body temperature regulation

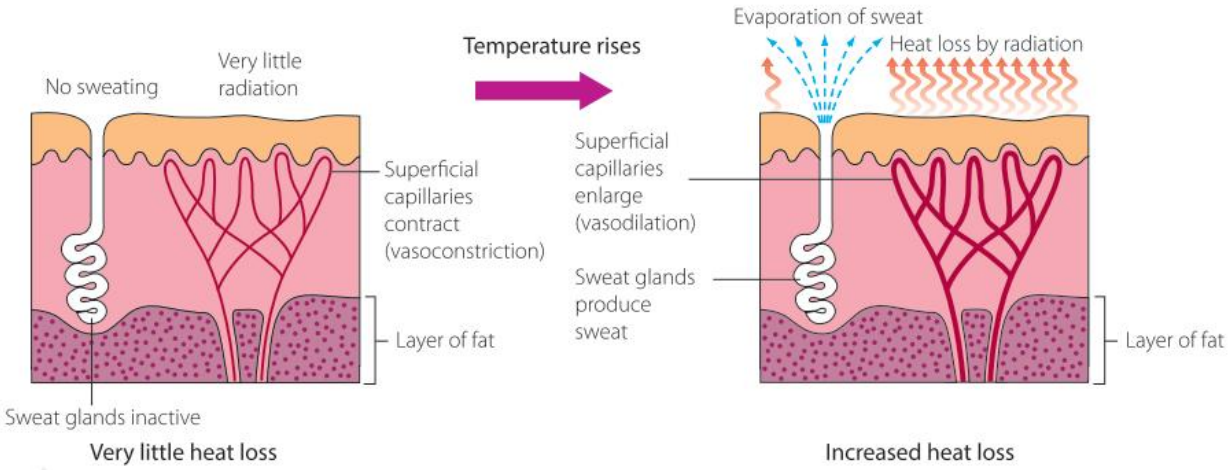

FIGURE 14.5 Vasodilation increases heat loss.

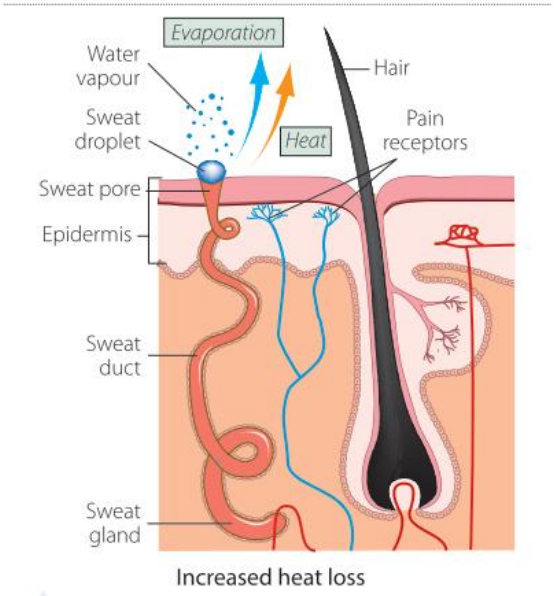

FIGURE 14.6 Evaporation of sweat brings about heat loss.

Warming the body

When receptors detect a decrease in the body temperature (stimulus), messages are sent to the posterior (back) area of the hypothalamus, which is the heat-gain control centre. Messages are sent via the nervous system to effectors that initiate processes to conserve heat and warm the body. These include:

- vasoconstriction, which involves the blood vessels constricting (narrowing), removing blood from the skin surface and conserving heat (Fig. 14.7)
- contraction of the hair erector cells, causing hair/fur to stand on end, which traps a layer of warm air around the body, reducing heat loss
- release of thyroid-stimulating hormone (TSH) by the pituitary gland under the direction of the hypothalamus, which causes the thyroid gland to increase the amount of the hormone thyroxin produced. This will increase the rate of metabolism, generating more heat in the body
- rapid contraction of muscles in a process called shivering, which generates heat in the body.

The combined effect of these processes is an increase in body temperature, which counteracts the stimulus and returns the temperature to its set point (or normal level).

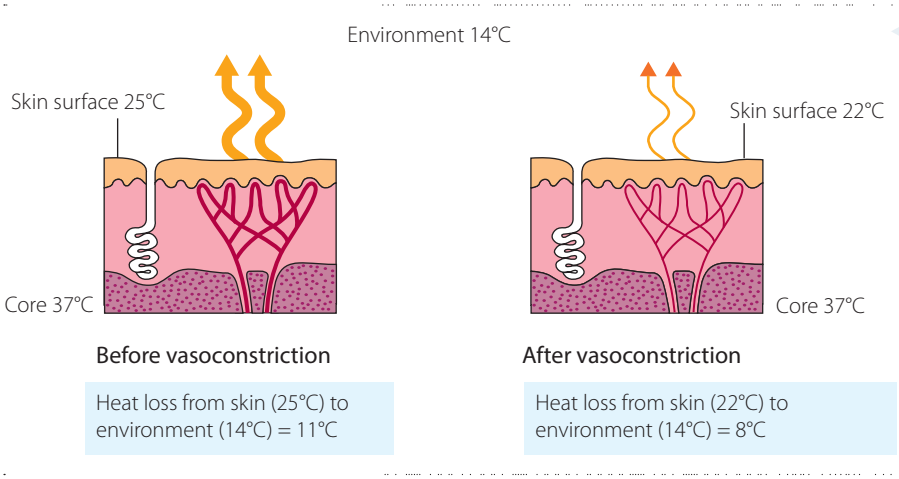

FIGURE 14.7 Vasoconstriction conserves heat by reducing heat loss from the skin to the environment.

Weblink
How body temperature is controlled

INVESTIGATION 14.1

A secondary source investigation into the regulation of glucose concentration in the blood

INTRODUCTION

The concentration of glucose in the blood must be maintained at a consistent level (set point) for the optimal metabolic functioning of the body. This concentration is regulated by a negative feedback mechanism, which can be represented in the form of a negative feedback loop.

The concentration of glucose is regulated by the hormones insulin and glucagon, which are produced in the pancreas. These hormones act on numerous tissues in the body.

Information and communication technology capability

Critical and creative thinking

AIMS

- To investigate the role of the pancreas in the regulation of glucose levels in the blood
- To construct a negative feedback loop that represents the regulation of glucose concentration in the blood

Weblink
Regulation of glucose levels in the blood

METHOD

1. Using the *background information* in the weblink as a starting point, research the processes involved in the regulation of blood glucose concentration.
2. Make sure you use a number of different sources and check the relevance, accuracy, validity and reliability of each source.
3. Answer the questions below to help guide your research.
4. Using the information you have gathered, construct a negative feedback loop to represent the processes involved in keeping the glucose concentration in the blood within tolerance levels.

Refer to Chapter 1, page 10 for guidance on to how to assess the relevance, accuracy, validity and reliability of sources.

QUESTIONS

1. Outline the importance of glucose in the human body.
2. Suggest when the concentration of glucose in the blood could:
   a. increase
   b. decrease.
3. Describe what could happen to the body if the blood glucose concentration became:
   a. too high
   b. too low.

4 Identify the two hormones involved in the regulation of the blood glucose concentration.

5 Where are the receptors for detecting the concentration of glucose in the blood located?

6 Describe the processes that occur to bring the blood glucose concentration back to its set point when it is:

- **a** too high
- **b** too low.

7 Copy and complete the table below.

FACTOR	GLUCOSE CONCENTRATION IN THE BLOOD	
	TOO HIGH	TOO LOW
Stimulus		
Receptor		
Control centre		
Effectors		
Response		
Result		

RESULTS

1 Construct a negative feedback loop to represent the regulation of blood glucose concentration.

2 Describe the processes involved in the regulation of blood glucose concentration.

DISCUSSION

Explain the importance of:

- **a** regulating the concentration of glucose in the blood
- **b** maintaining homeostasis in the body of an organism.

Weblink
Homeostasis of different conditions in the body

CONCLUSION

Write summary sentences related to the aims of this investigation.

CHECK YOUR UNDERSTANDING 14.1

1 Define 'homeostasis'.

2 **a** Identify the two systems involved in the control and coordination of homeostasis and the form in which messages are carried in each system.

b How are the messages carried in each system?

3 Explain why homeostasis is important.

4 Sketch a graph that indicates how an internal condition (such as body temperature) can vary about a set point within upper and lower tolerance limits.

5 **a** What are the two main stages of homeostasis?

b What is the role of the control centre in homeostasis?

c Identify an important control centre in the brain.

6 Define 'negative feedback mechanism'.

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7 Copy and complete the following table using the information in the negative feedback loop shown in Figure 14.4.

COMPONENT OF MECHANISM	BODY TEMPERATURE	
	TOO HIGH	TOO LOW
Stimulus		
Receptor		
Control centre		
Effectors		
Response		
Feedback		
Result		

8 The negative feedback mechanism shown in Figure 14.8 controls the level of oxygen (O_2) in the blood.

a Identify the following components of this negative feedback mechanism from the model shown:

- **i** stimulus
- **ii** receptor
- **iii** control centre
- **iv** effector
- **v** response
- **vi** feedback.

b Describe how the levels of oxygen in the blood are controlled.

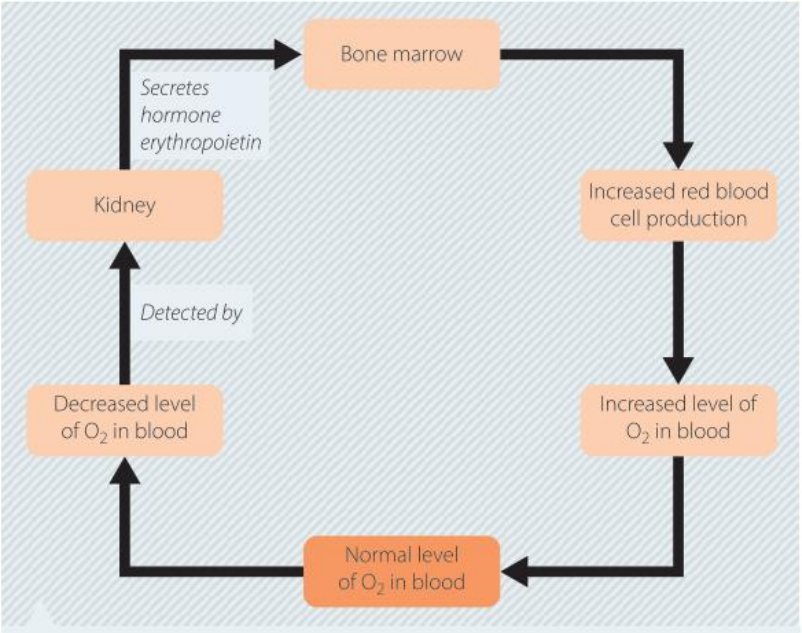

FIGURE 14.8 Negative feedback mechanism controlling the level of O_2 in the blood

14.2 Mechanisms to maintain homeostasis

Numerous mechanisms are used by organisms to maintain their internal environment within tolerance limits. These include adaptations of various types as well as internal coordination systems, such as the nervous and endocrine systems. All these systems play an important role in the maintenance of homeostasis, to allow the organism to achieve optimal metabolic efficiency, which contributes to the health and wellbeing of the organism.

Internal coordination systems

The nervous and endocrine systems are internal systems that work together to ensure that homeostasis is maintained. The role of these systems is to coordinate and provide pathways of communication for the negative feedback systems that operate within the body to maintain homeostasis. The two systems work together or individually to provide a pathway for relaying messages via nerves or hormones, from the receptors to the control centre and then on to the effectors to initiate a response, which will counteract the stimulus and ensure that the internal condition remains within tolerance limits.

The transport of messages in each system is very different. Nerve impulses are transported very quickly along nerves to specific locations in the body. Messages sent using hormones are in the form of chemical substances that are transferred to regions of the body in the bloodstream at a much slower rate than the transmission of nerve impulses.

Receptors

In both systems, receptors are responsible for detecting stimuli in the form of any changes from the set point that are outside the tolerance limits. Receptors contain sensory cells and can take numerous forms depending on the stimuli that activate them.

In their more complex form, receptors are concentrated in particular areas, forming **sense organs** such as the eye, the ear and the tongue. In many animals, including humans, receptors in sense organs detect stimuli in the *external* environment.

Interoceptors are receptors within the body that detect internal stimuli related to homeostasis.

Receptors may be named according to the type of energy or molecules they detect.

The receptors that are important in homeostasis are as follows.

- **thermoreceptors** detect changes in temperature. Thermoreceptors in the skin are nerve endings that are sensitive to heat or cold and send information to the brain about the external temperature. Internal thermoreceptors in the hypothalamus detect the temperature of the blood as it flows through the brain
- **chemoreceptors** detect the concentration of certain chemicals inside the body. Chemoreceptors are located in certain blood vessels and detect the pH as well as levels of certain chemicals, such as carbon dioxide and oxygen
- **osmoreceptors** detect changes in osmotic pressure and are located in the hypothalamus. Osmotic pressure in the blood is determined by the concentration of substances dissolved in the blood plasma. Small changes in osmotic pressure cause the body to implement processes that regulate the amount of water in the body, keeping it within the tolerance limits.

KEY CONCEPTS

- The nervous and endocrine systems coordinate and provide communication pathways to maintain homeostasis.
- In the nervous system, nerve impulses are transported very quickly along nerves to specific locations in the body.
- In the endocrine system, glands produce hormones in the form of chemical substances that are transferred to regions of the body in the bloodstream at a much slower rate than the transmission of nerve impulses.
- Receptors detect stimuli.
- External receptors are often grouped together in sense organs.
- Internal receptors (interoceptors) detect internal stimuli.

TYPE OF RECEPTOR	STIMULI DETECTED
Thermoreceptor	Changes in temperature
Chemoreceptor	Changes in pH and concentration of chemicals
Osmoreceptor	Changes in osmotic pressure

The nervous system

The neural pathways by which messages travel in the body are provided by the nervous system. As well as providing a communication pathway, the nervous system acts as a control centre to coordinate activities that maintain homeostasis within the body.

The nervous system has two main parts: the central nervous system (CNS) and the peripheral nervous system (PNS) (Fig. 14.9).

The CNS is composed of the brain and spinal cord; the PNS comprises all other nerves throughout the body that are not part of the CNS.

The peripheral nerves carry information to and from the CNS. The information carried by nerves consists of 'messages' transmitted in the form of electrochemical impulses.

Some actions involving the nervous system may take place voluntarily, but all those involved in homeostasis take place without conscious thought. They are involuntary and many are innate, unconditioned reflexes in response to a particular stimulus.

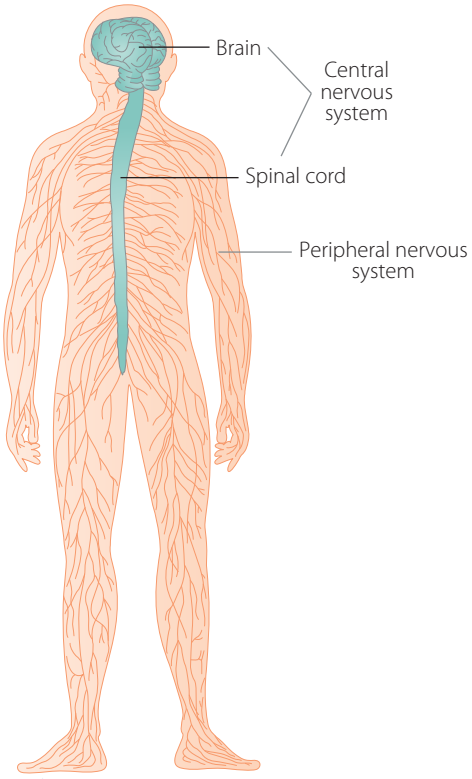

FIGURE 14.9 The two main parts of the nervous system: the central nervous system and the peripheral nervous system

Neurons

The millions of units that make up the nervous system are called nerve cells or neurons. Although no two neurons are exactly alike in size, shape and function, they all contain three common structures (Fig. 14.10):

- a cell body that contains a nucleus and many of the organelles found in other cells. These form the 'grey matter' of the CNS
- one or more fine branching extensions, called dendrites, that are extensions of the cytoplasm of the cell body. Dendrites receive messages in the form of impulses from other axons and conduct these nerve impulses *towards* the cell body. In sensory neurons the single, elongated dendrite is called a dendron
- one single, very long extension of the cytoplasm of the cell body, called an axon. Axons conduct messages *away from* the cell body and form the 'white matter' of the CNS.

Weblink
Different parts of the nervous system

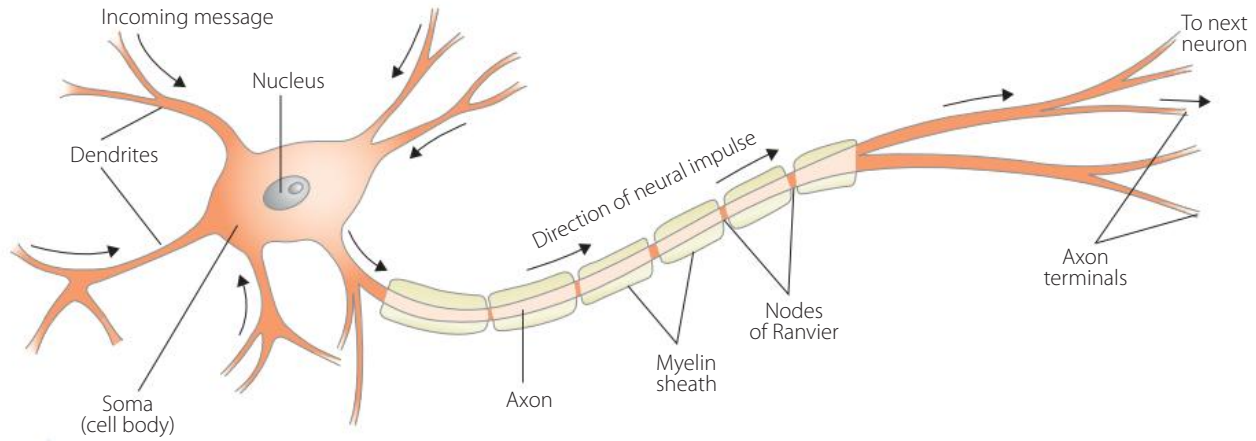

FIGURE 14.10 Structure of a typical neuron

Neurons are classified according to their function and the direction in which the nerve impulses are carried.

- **Sensory neurons** carry impulses from the sensory cells in the peripheral nervous system to the CNS. They usually have the cell body at the side, one long dendron and short axons.
- **Motor neurons** transfer messages from the CNS to effectors such as muscles or glands. The dendrites are usually short and the axon quite long.
- **Interneurons** (also known as association or connector neurons) are located within the CNS and are the link between the sensory and motor neurons. They have short dendrites and either long or short axons.

The three major types of neurons – sensory, motor and interneurons – are shown in Figure 14.11.

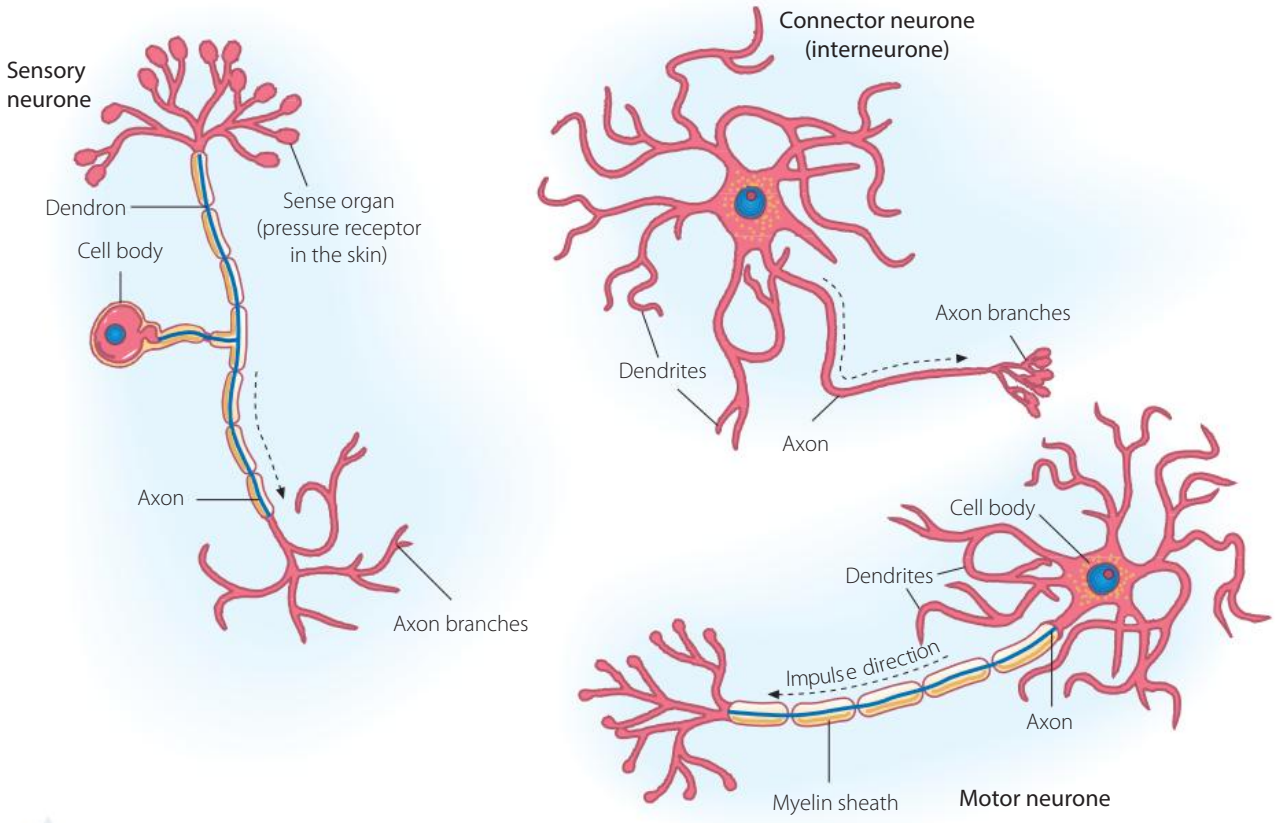

FIGURE 14.11 The structure of the three major types of neurons: sensory, motor and connector (interneurons)

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When a nervous impulse is transferred from the axon of one neuron to the dendrites of the adjacent neuron it must cross a small gap, or **synapse**, as the adjacent neurons do not actually touch (Fig. 14.12).

The fibres of the neurons are gathered into bundles held together by a connective tissue sheath to form the nerves (Fig. 14.13). This provides a structured pathway for the transmission of electrochemical impulses along the axons of the neurons.

The messages transmitted by neurons as nerve impulses are in the form of electrochemical impulses. This is the quickest way to initiate responses by the body to stimuli in order to coordinate and maintain homeostasis.

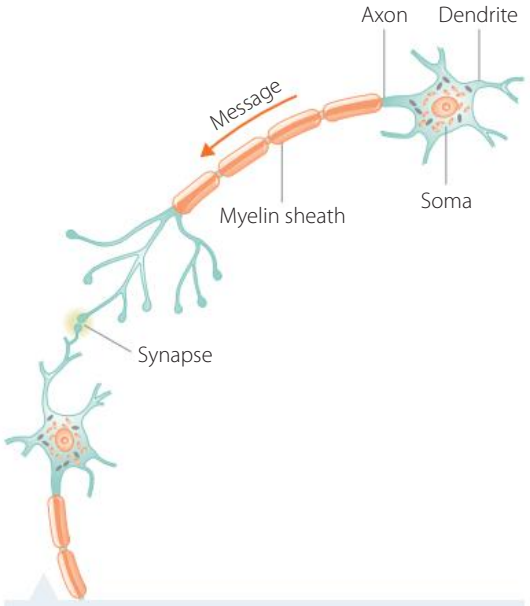
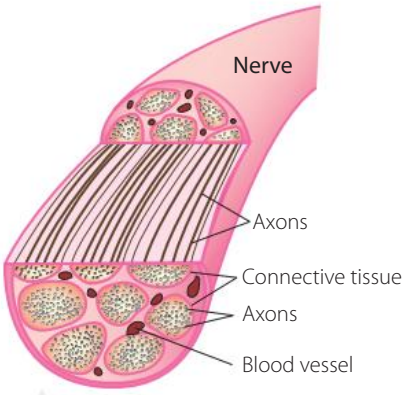


FIGURE 14.12 The gap between the axon terminals and dendrites of adjacent neurons is called a synapse.



Source: Purves et al., Life: The Science of Biology, 4th Edition, by Sinauer Associates (www.sinauer.com) and WH Freeman (www.whfreeman.com)

FIGURE 14.13 A nerve contains many neurons bundled together.

Transmission of nerve impulses – the action potential

Electrochemical impulses involve a change in the electrical potential of the cell membrane of the axon – this temporary change is known as an **action potential**. This is brought about by a change in the concentration of electrically charged chemicals, called **ions**, on either side of the cell membrane of the axon. In other words, the change in the concentration of the chemicals by movement across the cell membrane causes an electrical impulse.

Sodium ions (Na^+), potassium ions (K^+) and chloride ions (Cl^-) are important for the transmission of electrochemical messages in the nervous system. These ions are on both sides of the cell membrane, both inside and outside the cell, but their concentrations differ. Also inside the cell are large negatively charged organic ions (M^-).

A factor that has an influence on the movement of these ions across the cell membrane is that the cell membrane is selectively permeable. It allows some substances to pass through easily, such as K^+ ions, but hinders the passage of Cl^- and Na^+ ions. The large M^- ions are unable to move through the cell membrane.

At rest

A neuron is said to be at rest if it is not transmitting any electrochemical messages. In this state the ions inside and outside the cell attempt to balance themselves out. This is not possible, as Na^+ ions can only move through the cell membrane of the neuron through special ion channels, which are closed when the neuron is at rest. There are a large number of Na^+ ions outside the cell compared to the number of K^+ ions inside the cell. Also, there are a large number of organic M^- ions trapped inside the cell.

Because of this there are more total negative charges on the inside of the cell than on the outside, and there is said to be a potential difference across the membrane. The value of this potential difference, called the *resting membrane potential*, is –70 mV (millivolts). The inside of the membrane is said to be negative in relation to the outside of the membrane – the membrane is **polarised** (Fig. 14.14).

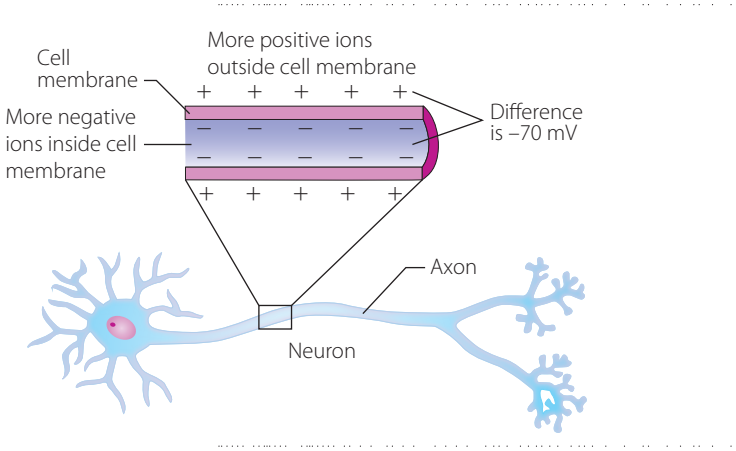


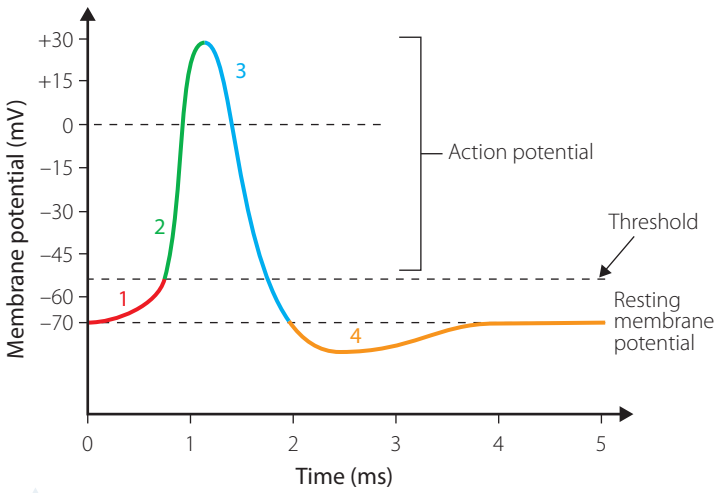
FIGURE 14.14 The resting potential of the axon cell membrane is –70 mV. The membrane is polarised.

Action potential – depolarisation and repolarisation

When a stimulus is detected by a neuron, it causes sodium channels in the cell membrane to open. Because there are many more Na^+ ions outside the neuron than inside, the Na^+ ions move into the neuron and reduce the overall negative charge. If the stimulus is 'strong' enough to open enough Na channels to allow Na^+ ions to move into the neuron and change the resting potential to its *threshold* value of –55 mV, the movement of Na^+ ions into the neuron will continue independently of the stimulus. This causes the inside of the membrane to become more positive in relation to the outside of the neuron, and the potential to move to 0 mV and beyond. The membrane has been *depolarised*. Just as quickly, the potassium channels open and K^+ ions move out of the neuron, causing the *repolarisation* of the membrane. The potassium channels stay open a little longer and the action potential goes past –70 mV (hyperpolarisation) before returning to its original resting state. This rapid depolarisation and repolarisation is called the *action potential* (Fig. 14.15).

Weblink
Watch these animations and summarise the steps involved in an action potential.

Weblink
Use the interactive to deepen your understanding of the way in which action potentials are formed and how nerve impulses move along neurons.



1 Slow depolarisation of the membrane brings the potential to the threshold.

2 Sodium channels in the membrane open; sodium ions flood into the cell; membrane becomes depolarised; membrane voltage rises.

3 Sodium channels close and membrane becomes repolarised.

4 Membrane returns to resting state.

FIGURE 14.15 Graph showing an action potential

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The action potential is only activated if the stimulus is strong enough to cause the potential to reach a threshold value of –55 mV. Once this value is reached, the action potential goes ahead automatically. If the threshold value is not reached, there is no action potential and therefore no nerve impulse is generated. Either the threshold potential is not reached or a full action potential is fired. This is the 'all or none' principle.

Each action potential causes another action potential in the neighbouring region of the neuron. This series of action potentials along the neuron is the nerve impulse (Fig. 14.16).

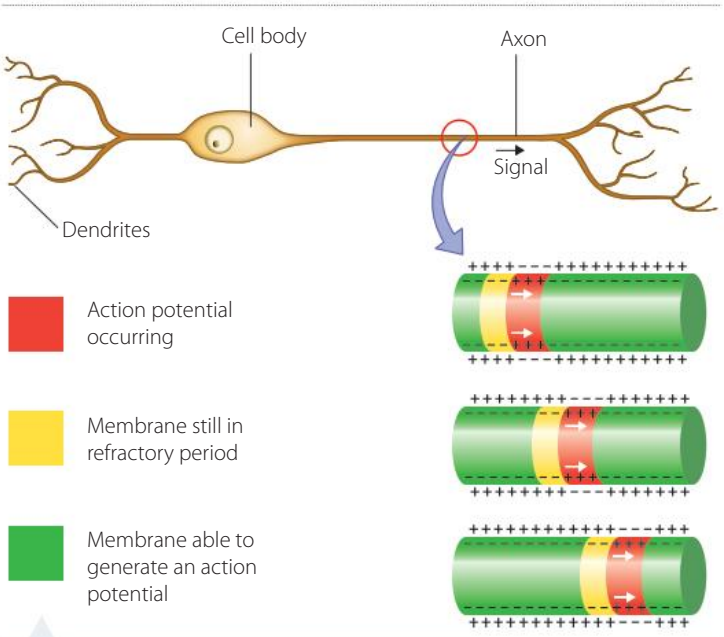

FIGURE 14.16 Transmission of a nerve impulse along an axon

Bridging the gap: synapse

When a nerve impulse reaches the axon terminal, it has to move across a small gap, the synapse, to the dendrites of the next neuron (Fig. 14.17). The action potential triggers the release of chemicals, called neurotransmitters, from the synaptic vesicles. These chemicals move across the synapse to receptors in the dendrites in the adjacent neuron, initiating an action potential in order to continue the nerve impulse.

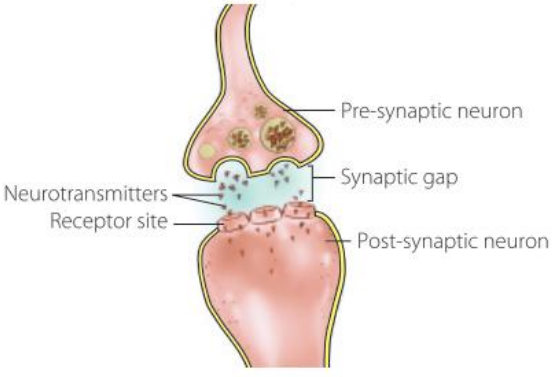

FIGURE 14.17 Neurotransmitters transfer nerve impulses across the synapse.

Weblink
Synapses
Watch the animation and create a flow chart to show how electrical impulses are transferred from one neuron to the next.

INVESTIGATION 14.2

A secondary-source investigation to graph action potential

INTRODUCTION

Numeracy

Action potential is important for the movement of electrochemical impulses along a neuron. A series of action potentials along a neuron is what makes up a nerve impulse. In this investigation you will be required to graph the provided values for the change in electrical potential between the inside and outside of the cell membrane of the axon of a neuron. These values are recorded as an action potential is generated and goes to completion before being stimulated in the neighbouring region.

AIM

To graph an action potential

METHOD

1. Use the information provided in Table 14.1 to draw a line graph of the membrane potential against time.
2. Label the following areas on the graph: resting potential, threshold, depolarisation, maximum action potential, repolarisation, hyperpolarisation and resting potential at the end.

TABLE 14.1 Membrane potential measured against time

MEMBRANE POTENTIAL (mV)	TIME (ms)	MEMBRANE POTENTIAL (mV)	TIME (ms)
–70	0	–76	2.25
–70	0.25	–74	2.5
–65	0.5	–72	2.75
–50	0.75	–71	3.0
+10	1.0	–70	3.25
+34	1.25	–70	3.5
–50	1.5	–70	3.75
–60	1.75	–70	4.0
–80	2.0		

3. Explain what is happening at each of the labelled stages. This can be presented in a table, as a dot point summary or in any other appropriate fashion.

RESULTS

1. Draw a line graph from the information presented in the table.
2. Provide a summary of what is happening at each of the labelled stages.

DISCUSSION

1. Look at the graphs in Figure 14.18, which shows two stimuli of different strengths changing the membrane potential of the neuron.
   Is an action potential generated in both (a) and (b)? Justify your answer.
2. Does a stronger stimulus cause a stronger action potential? Explain why or why not.

CONCLUSION

Write a summary sentence to address the aim of this investigation.

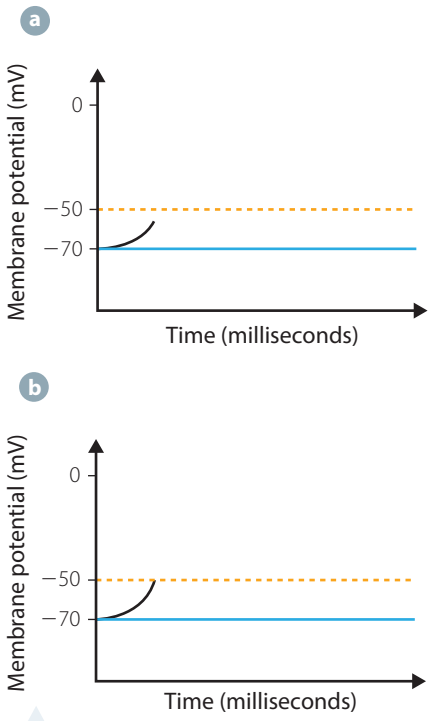

FIGURE 14.18 Change in membrane potential in response to stimuli **a** and **b**

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INVESTIGATION 14.3

A first-hand and secondary-source investigation to investigate the structure of neurons, and to observe and model neurons

Information and communication technology capability
Literacy
Critical and creative thinking
Personal and social capability

INTRODUCTION

The nervous system is an integral part of the communication system in the body. Neurons that make up the nerves have a structure that is specifically suited to their function. In this investigation you will research the function of the structures in a typical neuron and observe the structure of neurons both directly and indirectly. You will also construct a model of a neuron to be reviewed by your peers.

AIMS

- To provide a written description of the function of the different parts of a typical neuron
- To observe the structure of neurons
- To construct a model of a neuron

MATERIALS

- access to reference books and the Internet
- digital camera
- monocular microscopes
- prepared slides of the following:
  - cross-section of spinal cord
  - motor neuron
  - brain tissue
- materials to construct a model of a neuron; this could include, but is not limited to, the following:
  - coloured cardboard
  - straws
  - thin foam-like material
  - wool
  - small pasta tubes
  - pipe cleaners
  - coloured paper
  - spherical foam balls
  - plastic tubing of different thicknesses
  - glue
  - scissors
  - knife

Weblink
3D model of a typical neuron

RISK ASSESSMENT

RISK ASSESSMENT

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?
Knife	Sharp edges can cause cuts.	Use knife with care, hold by the handle and keep fingers away from sharp edge of knife.
Microscope slides/ coverslips	Sharp edges can cause injury if broken.	Handle with care. Push gently on coverslip. Always focus by moving objective lens away from slide.

METHOD

A Research

1. The basic structure of a neuron has been covered on page 480 and is shown in Figure 14.10.
2. Refer to Figure 14.10 and note all the labelled areas.
3. Research numerous sources in order to outline any distinguishing features of, and the function of, each of the labelled structures.
4. Collate the information you have found and present it in an appropriate written form, such as a dot-pointed summary or a table.
5. Research using the Internet and other secondary sources to find labelled images of neurons seen through the light microscope and electron micrographs of neurons, similar to the ones shown in this chapter.
6. Access the website shown in the weblink or similar, to view a 3D model of a typical neuron.

»

B Observing neurons

1. Obtain a microscope and the prepared slides supplied.
2. Following the correct microscope procedures, focus each of the slides provided, first on low power and then on high power.
3. Observe any neurons present and identify any of the structures visible with the help of the images you have researched in Step 5 above.
4. If possible, take a photo of one the neurons observed using your mobile phone, print it out and label the structures you identified.
5. If it is not possible to take a photo, draw a labelled scientific diagram of one the neurons you observed.

Refer to Chapter 1 page 10 for guidance on to how to assess the relevance, accuracy, validity and reliability of sources.

C Making a model

1. Working in pairs, design a model of a motor neuron.
2. This model could take any form – it could be 2D, using cardboard as a base with different materials representing each of the structures present in a neuron, or it could be a 3D structure.
3. The different parts of the model should be identified, either with labels or with a key.
4. The materials you use for your model should be easily obtainable.
5. When your model is complete, submit it for peer review in a manner determined by your teacher.

RESULTS

Your results should include:

- information about the structures and functions of the structures in a typical neuron, recorded in a dot point summary, table or other appropriate form, as instructed in the steps above
- a labelled photo or scientific diagram of one of the neurons you observed using the light microscope
- a completed model, including labels, of a motor neuron.

DISCUSSION

1. Assess the relevance, accuracy, validity and reliability of the secondary sources you used to find the required information about neurons.
2. Suggest any improvements that could have been made to this investigation.
3. **a** Outline why scientists use models.
   b Describe how constructing a model of a motor neuron improved your understanding of neurons.

CONCLUSION

Write a few summary sentences to address the aims of this investigation.

KEY CONCEPTS

- The two main parts of the nervous system are the central nervous system (CNS) and the peripheral nervous system (PNS).
- The nervous system contains millions of neurons (nerve cells), which transmit messages via electrochemical impulses.
- A typical neuron contains a cell body, dendrites and an axon.
- The three types of neurons are sensory neurons, motor neurons and interneurons (connector neurons).
- The synapse is the small gap between the axon terminals and dendritic terminals of adjacent neurons.
- Nerves are made up of bundles of neuronal fibres.
- A stimulus will cause a change in ion concentrations across the cell membrane of the axon, which in turn alters the membrane potential.
- If the membrane potential reaches the threshold value, an action potential involving depolarisation, and then repolarisation, is instigated.
- Each action potential causes another action potential in the next region of the neuron. This series of action potentials along the neuron is the nerve impulse.

- If the threshold value is not reached, there will be no action potential and therefore no nerve impulse generated.
- Neurotransmitters released at the axon terminal transfer the 'message' across the synapse to the adjacent neuron and stimulate an action potential in the dendrites of the receiving neuron.

CHECK YOUR UNDERSTANDING 14.2a

1 **a** What is the role of receptors in the body?
   b Name three types of receptors and describe the function of each.
2 **a** Name the two main parts of the nervous system.
   b Outline the structure and function of each part.
3 **a** Sketch a diagram of a typical neuron.
   b Label the dendrites, cell body, nucleus, axon, myelin sheath and nodes of Ranvier.
   c Draw an arrow on your diagram to indicate the pathway of the nerve impulses along the neuron.
4 Construct a table that summarises the three types of neurons and their functions.
5 Identify the form that messages take when travelling along neurons.
6 Summarise the steps involved in the passage of a nerve impulse along a neuron. This could take the form of a flow chart.
7 Name the small gap between adjacent neurons, and outline how nerve impulses move across this gap.

The central nervous system

The central nervous system (CNS) is made up of the brain and the spinal cord, and is integral to the maintenance of homeostasis. Both the brain and the spinal cord are made up of two types of nervous tissue: grey matter and white matter. Grey matter consists mainly of neuron cell bodies, while white matter is nerve fibres surrounded by myelin sheaths, which cause the white appearance. In the brain, the grey matter tends to be on the outside, whereas in the spinal cord it is in the centre.

The brain

The brain is the main control centre of the body and is therefore a very complex organ. It consists of numerous parts that all work together to ensure the efficient functioning of the body. The brain largely controls the maintenance of homeostasis.

The major parts of the brain are shown in Figure 14.19.

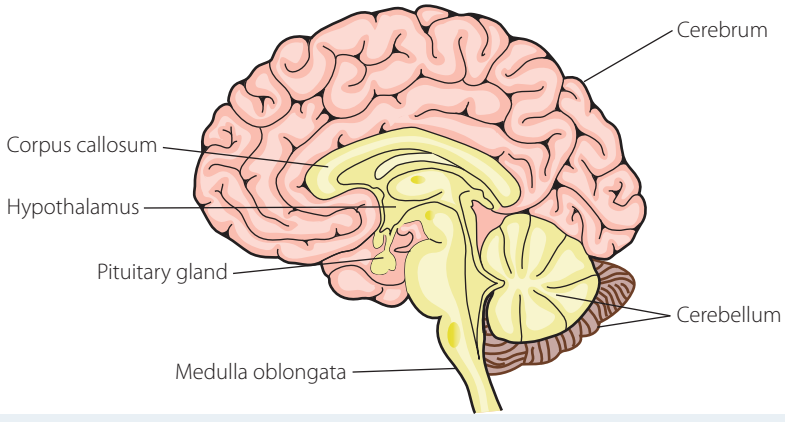


FIGURE 14.19 Cross-section of the brain, showing the major regions

The endocrine system is discussed on pages 488–492.

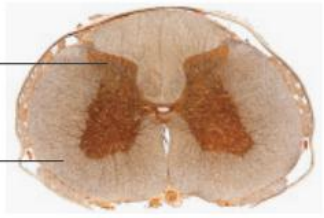
The **hypothalamus** is a small area of the brain that is located centrally and close to the pituitary gland. It is the control centre for the regulation of the many activities in the body that are required to maintain a stable internal environment (homeostasis). It achieves this by directing effectors to carry out a response either by sending messages via neural pathways or by chemical messages (hormones). Some of the conditions that need to be regulated are heart rate, body temperature, blood pressure, and the concentration of oxygen and carbon dioxide in the blood.

The hypothalamus is also the main link between the nervous system and the endocrine system. It is responsible for hormone secretions and directs the actions of the pituitary gland in coordinating other glands to secrete hormones in order to maintain homeostasis.

Weblink
An interactive look at the brain

The spinal cord

The spinal cord extends from the medulla oblongata down through the vertebral column to the waist area (Fig. 14.9). It contains the nerve fibres that provide the link for the pathway of nerve impulses between the peripheral nervous system and the brain (Fig. 14.20).



Manfred Kage/Science Photo Library

FIGURE 14.20 Cross-section of spinal cord

The two main functions of the spinal cord are:

- to act as a conduction pathway for nerve impulses from the receptors around the body to the brain, and for nerve impulses from the brain to the effectors. This is essential for the efficient functioning of all areas of the body, including the maintenance of homeostasis
- to coordinate reflex actions, such as removing your hand quickly when you touch something hot, before you feel the pain.

Weblink
Virtual dissection

Worksheet
Brain dissection

KEY CONCEPTS

- The CNS is made up of the brain and the spinal cord, and acts as the major control and coordination area for the maintenance of homeostasis.
- The main regions of the brain are the cerebrum, the cerebellum and the medulla oblongata.
- The corpus callosum provides a pathway for messages between the two sides of the brain.
- The hypothalamus provides a link between the nervous and endocrine systems, to assist in the maintenance of homeostasis.
- The spinal cord provides a pathway for nervous impulses between the brain and the rest of the body.

CHECK YOUR UNDERSTANDING 14.2b

1 Identify the areas of the brain indicated in Figure 14.21.

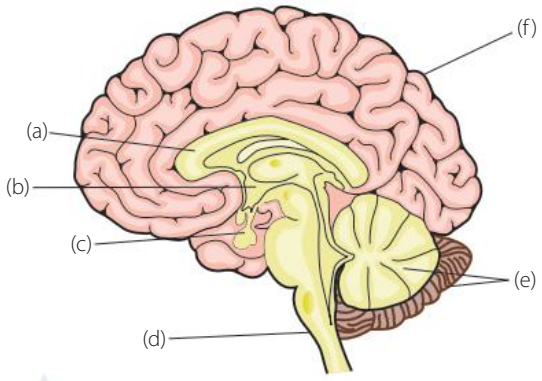


FIGURE 14.21 Major regions of the brain

2 Describe the two main functions of the spinal cord.

3 Compare the distribution of white and grey matter in the brain and spinal cord.

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The endocrine system

The endocrine system, along with the nervous system, regulates the activity of the body. Its main components are **hormones**, which are chemical messenger molecules secreted by **endocrine glands**. The word 'endocrine' comes from *endo* meaning 'from within' and *krine* meaning 'to secrete'.

These hormones are transported by the bloodstream to cells possessing the receptors for the particular hormone. The hormones cause these cells to change their activity in a way that will maintain homeostasis within the body. Hormones achieve this by influencing the activity of particular enzymes or the concentration of these enzymes in the cells, known as **target cells**. Figure 14.22 shows the location of the major glands of the endocrine system and the hormones they produce.

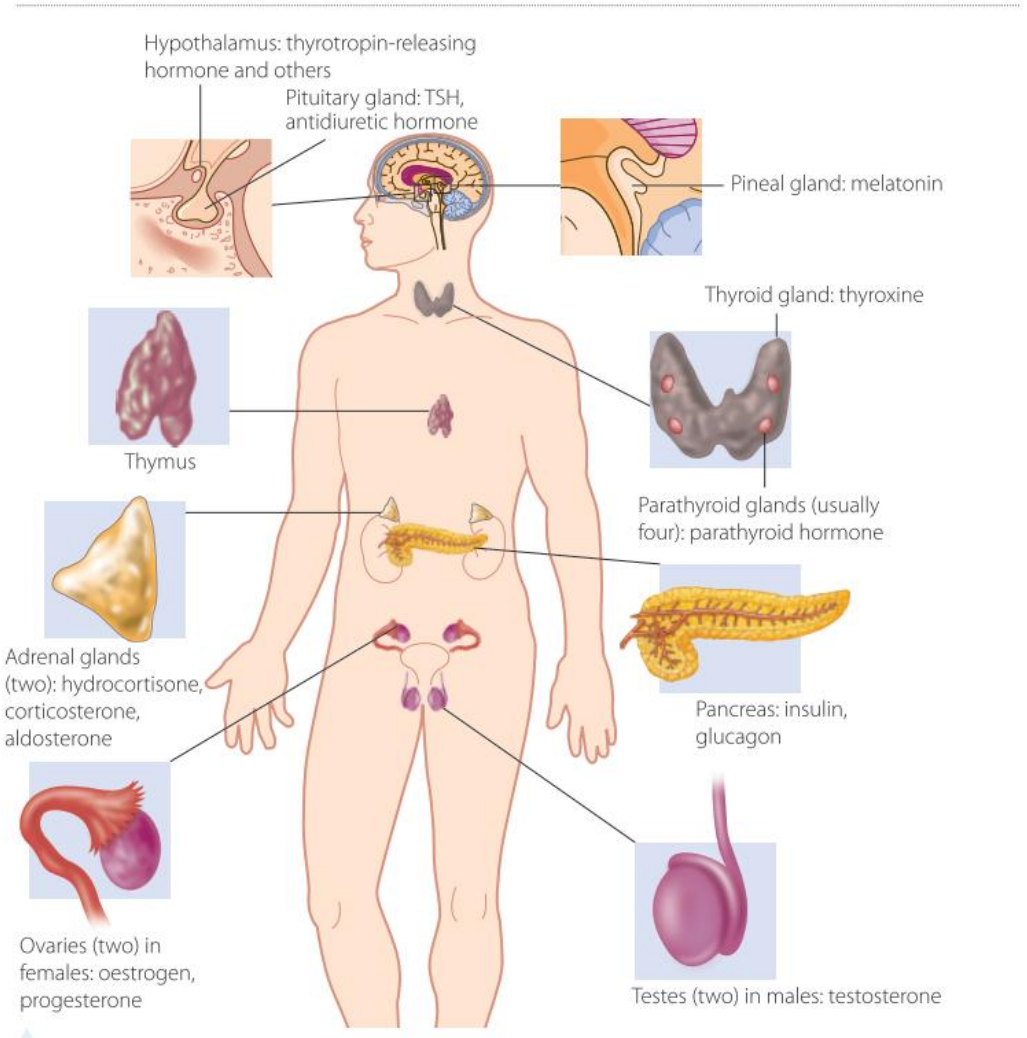

FIGURE 14.22 The location, hormones produced and function of the major glands of the endocrine system

Glands can be stimulated to secrete hormones by messages from the nervous system, by other hormones or by receptors located in the particular gland.

Pituitary gland

The **pituitary gland**, situated just below and working in close collaboration with the hypothalamus, is often referred to as the master gland. It releases hormones, often on direction from the hypothalamus, to regulate the activity of other glands.

Worksheet
Hormonal regulation of homeostasis

Weblink
Glands in the endocrine system
Create a table to summarise the characteristics of each gland in the endocrine system: location, hormone(s) produced and effect of hormone(s)

There are two distinct regions of the pituitary gland: the front (anterior) and the back (posterior) (Fig. 14.23). Hormones released by the hypothalamus control the anterior area of the pituitary gland, while the posterior section is controlled by nerve impulses.

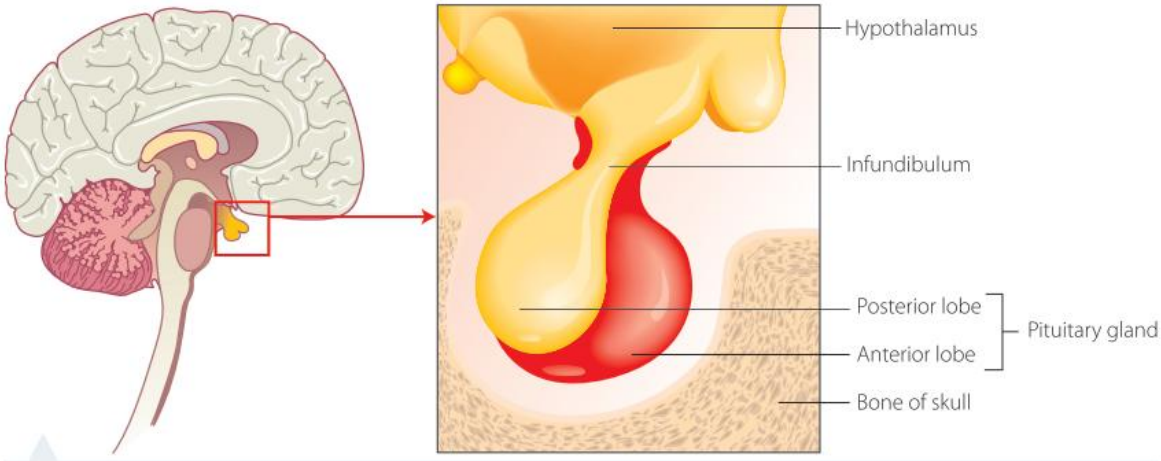

FIGURE 14.23 The pituitary gland is closely connected to the hypothalamus.

One hormone secreted by the anterior section controls growth. Other hormones secreted act on other glands to control the activity of the thyroid, the adrenal gland and the gonads (ovaries and testes).

One of the hormones secreted by the posterior section of the pituitary gland is antidiuretic hormone (ADH), which helps to regulate the concentration of water in the body. If receptor cells in the hypothalamus detect that the levels of water in the body are too low, the hypothalamus stimulates the pituitary gland to release ADH. This acts to conserve water in the body by promoting its reabsorption by the kidney tubules. The opposite occurs when the level of water in the blood is too high – the hypothalamus detects this and directs the pituitary gland to reduce its production of ADH. This leads to less water being absorbed in the kidneys and the increased excretion of water.

Weblink
Production and action of ADH

The following glands and the hormones they produce are primarily involved in the maintenance of further stable internal conditions (homeostasis) within the body.

Thyroid and parathyroid glands

The **thyroid gland** consists of two lobes located on either side of the neck (Fig. 14.24). The follicles in this gland predominantly produce an iodine-containing hormone called **thyroxine**. The amount of thyroxine produced is controlled by thyroid-stimulating hormone, which is released by the anterior pituitary gland.

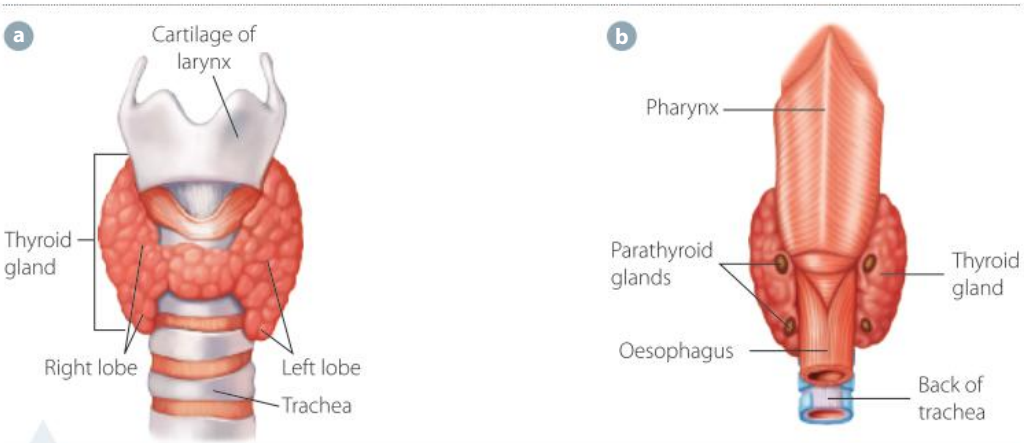

FIGURE 14.24 The location of the **a** thyroid and **b** parathyroid glands

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The release of thyroxine is in turn controlled by the hypothalamus when it releases a regulatory hormone into the pituitary gland. One of the reasons this may occur is that receptors in the hypothalamus detect a drop in the body temperature to below the tolerance limit.

Thyroxine is the means by which the message is transferred to the cells to increase their metabolic rate. In this process, energy is released for a number of purposes, one of which is to provide heat to maintain body temperature.

The **parathyroid gland** is made up of a number of small glands embedded in the surface of the thyroid gland (Fig. 14.24b). Its function is to maintain the level of calcium in the blood. Calcium is required for the successful transfer of nerve impulses in the nervous system and allows muscles to contract properly. The parathyroid gland monitors the blood flowing through it. When it detects that the calcium level is too low, it secretes **parathyroid hormone** (PTH). This hormone travels to the following effectors:

- bones – release calcium into the blood
- small intestines – absorb more calcium from digested food
- kidneys – reabsorb more calcium in the tubules.

The net result of this is to increase calcium levels, thus returning them to normal levels.

If the calcium levels detected are too high, the parathyroid gland stops secreting PTH.

Adrenal glands

The **adrenal glands** are located on the top of each kidney (Fig. 14.25) and are made up of two distinct parts. The outer portion of the adrenal gland is known as the **adrenal cortex** and the inner region is the **adrenal medulla**. These regions have very different functions and secrete different hormones.

The hypothalamus regulates activities in both regions. Some of the hormones produced by the cortex are regulated by negative feedback involving the hypothalamus and hormones produced by the pituitary gland. The medulla is regulated by nerve impulses from the hypothalamus.

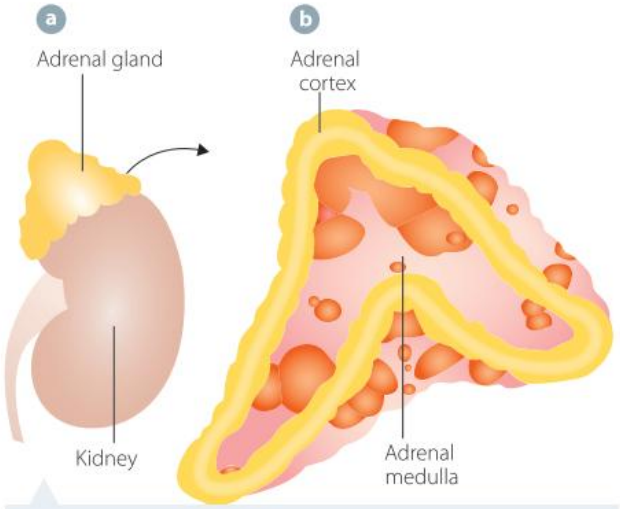

FIGURE 14.25 The adrenal glands are located at the top of the kidneys and have two parts – the adrenal medulla and the adrenal cortex. **a** external view of the kidney with adrenal gland; **b** longitudinal section through the adrenal gland

Adrenal cortex

When the hypothalamus is stimulated by receptors, it produces a hormone that in turn stimulates the pituitary gland to produce another hormone that stimulates the production of **hydrocortisone** (cortisol) and **corticosterone** by the cortex region of the adrenal glands.

Cortisol regulates how the body manages stress and how it converts carbohydrates, fats and proteins to energy. It also plays a role in regulating cardiovascular function and blood pressure.

Cortisol and corticosterone work together to regulate the immune response and suppress inflammatory reactions.

Another hormone secreted by the adrenal cortex is **aldosterone**, which increases the reabsorption of sodium ions and decreases the reabsorption of potassium ions in the kidney. If receptor cells in the kidneys detect low levels of sodium ions, they stimulate the adrenal cortex to produce more aldosterone, which causes greater reabsorption of sodium ions and decreased reabsorption of potassium ions. This results in sodium and potassium levels returning to values within their tolerance limits. The opposite occurs if the levels of sodium are too high and the levels of potassium are too low. The levels of sodium ions in the blood are linked to the maintenance of blood volume and blood pressure. A low level of sodium ions in the blood will reduce blood volume and therefore blood pressure.

Pancreas

The endocrine portion of the **pancreas** consists of structures called pancreatic islets or the **islets of Langerhans** (Fig. 14.26), where the hormones **insulin** and **glucagon** are produced. These islets contain two types of cells – alpha and beta cells.

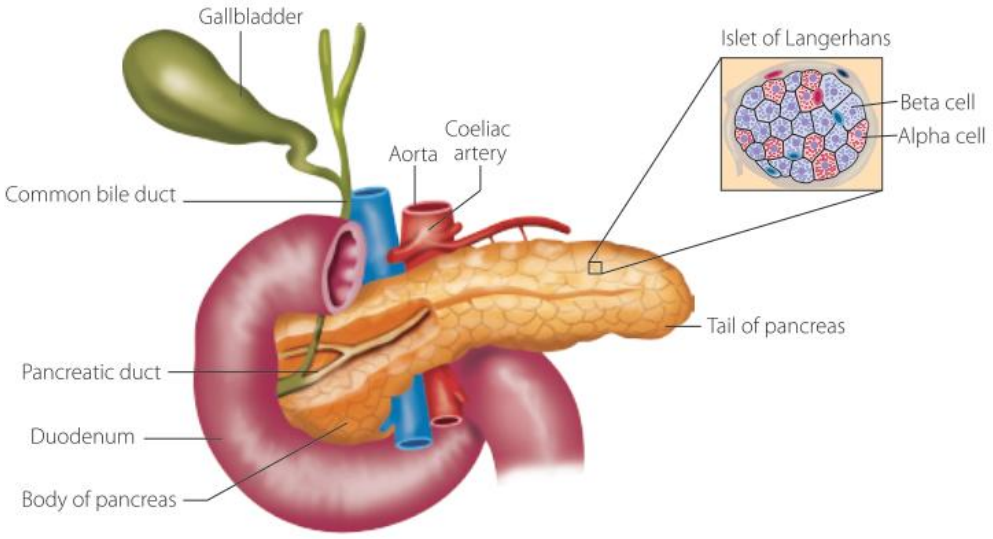

FIGURE 14.26 The pancreas, showing the alpha and beta cells in the islets of Langerhans

Chemoreceptors in the beta cells detect high levels of glucose in the blood and stimulate the production of insulin. Insulin causes glucose to be removed from the blood in a number of ways:

- In the liver, the glucose is converted into glycogen and fat.
- Skeletal muscles convert the glucose into glycogen.
- Glucose is converted into fat in fat storage tissue.

When the levels of glucose decrease, the production of insulin decreases.

Alpha cells in the islets of Langerhans produce glucagon in response to low levels of glucose in the blood. Glucagon causes the levels of glucose in the blood to increase by stimulating the production of glucose, by:

- the breakdown of glycogen in the liver
- the breakdown of fat in the liver and the fat storage tissues.

When the level of glucose increases back to normal, the production of glucagon is reduced.

KEY CONCEPTS

- The endocrine system assists in the regulation of homeostasis and has two main parts: the glands and the hormones they secrete.
- Hormones are transported by the bloodstream and cause cells to change their activity in a way that will maintain homeostasis.
- Glands can be stimulated to secrete hormones by messages from the nervous system, by other hormones or by receptors located in the particular gland.

CHECK YOUR UNDERSTANDING 14.2c

1. Name the two main components of the endocrine system.
2. Define 'target cells'.
3. Outline the different ways in which glands can be stimulated to secrete hormones.
4. Why is the pituitary gland often referred to as the 'master gland'?

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5 **a** Outline the relationship between the hypothalamus and the pituitary gland in maintaining homeostasis for certain internal conditions.

b Provide an example to demonstrate the interaction you described in **a**, in controlling the level of one named internal condition.

c Draw a labelled negative feedback loop to summarise the example you provided in **b**.

6 Describe the role of the hormones insulin and glucagon in maintaining the level of glucose in the blood.

7 Outline the relationship between the thyroid and parathyroid glands, and describe the contribution of each of these glands in maintaining homeostasis.

8 Predict the effect on the body of a disease called Addison's disease, which causes a decrease in the production of the hormones cortisol and aldosterone.

9 **a** Construct a feedback loop using the information provided on page 491 to summarise the control of calcium levels in the blood.

b Research the condition 'osteoporosis' and explain how this condition is related to the maintenance of calcium levels in the blood.

10 Discuss, using examples, how the endocrine system contributes to the maintenance of homeostasis.

14.3 Adaptations in endotherms

Endotherms are organisms that are able to maintain their body temperature within a very narrow range of tolerance limits despite variations in the **ambient** temperature (temperature of the environment). These organisms rely on internal sources such as metabolic activity in order to maintain their body temperature.

An **adaptation** is a characteristic that an organism possesses that will increase the survival and reproductive chances of that organism in its environment. Endotherms show a combination of adaptations to assist them in the regulation of body temperature, otherwise known as **thermoregulation**.

Endotherms that live in areas of high temperature need adaptations that reduce their exposure to heat and increase their ability to lose heat. Those that live in colder environments benefit from having adaptations that increase their ability to gain heat and reduce their heat loss.

Adaptations can be divided into three major groups:

- **behavioural** – the way an organism acts
- **structural** – the physical characteristics of the organism
- **physiological** – the way the organism's body functions.

Adaptations were discussed in *Biology in Focus Year 11*, Chapter 8.

Behavioural adaptations

One behavioural adaptation seen in endotherms to assist in thermoregulation is that they alter the position of their body and/or move to different areas to increase or decrease the amount of exposure of their surface area to the sunlight.

If the ambient temperature is too high, they may change the position of their body to reduce the surface area exposed, seek shade, shelter in burrows or move into water to cool down.

During the hottest part of the day, the red kangaroo (*Macropus rufus*) will seek or and sit in a position where its hind legs and tail are shaded by the rest of its body – they are positioned at right angles to the body, with the tail pointing forward, to reduce the large surface area exposed to the sun (Fig. 14.27a). Fairy penguins (*Eudyptula minor*) move into water to cool down. Many penguin species, including fairy penguins, huddle together in cold conditions to reduce the surface area of each penguin exposed to the cold and thus conserve heat (Fig. 14.27b). The mountain pygmy possum (Figure 14.30b) curls into a ball, drawing all appendages (legs, nose, ears and tail) in towards the body to reduce the surface area

exposed to the cold. They also use a burrow to shelter from the cold in shorter periods of low ambient temperature.

FIGURE 14.27 Behavioural adaptations to assist temperature regulation: **a** red kangaroos lying in the shade; **b** penguins huddling together for warmth

FIGURE 14.28 The bilby has a number of adaptations to assist in temperature regulation, including retreating into a burrow, nocturnal activity, high surface area to volume ratio and large, flat ears.

Nocturnal activity is another common behavioural adaptation to assist in regulating body temperature. It is seen in birds and mammals that live in habitats where the daytime temperature is very high. Nocturnal animals remain relatively inactive during the heat of the day, so that they do not generate additional metabolic body heat as result of increased activity.

An example is the bilby, *Macrotis lagotis*, which inhabits hot, dry regions of Australia (Fig. 14.28). The bilby shelters in its burrow during the day to avoid the heat, and comes out at night to forage for food.

Migration is another behavioural adaptation that assists in thermoregulation. Migrating organisms move to a different habitat that is within their tolerance range.

The grey plover (*Pluvialis squatarola*) (Fig. 14.29a) breeds in the northern hemisphere between May and August, then migrates to Australia during August and stays until April. This migration allows the birds to avoid the severe weather of winter. Many aquatic endotherms migrate from one area to another to avoid being exposed to extreme temperatures. Humpback whales (*Megaptera novaeangliae*) (Fig. 14.29b) in the southern hemisphere migrate annually from their southern feeding grounds to the warmer waters in the north to mate.

Structural adaptations

Structural adaptations that assist with temperature control include *insulation*, such as fur, hair and feathers, which trap a layer of air next to the skin, reducing the amount of heat lost. The feathers of fairy penguins provide an insulating layer to reduce the amount of heat lost. This layer of air can be altered depending on the ambient temperature. In cold conditions the feathers are lifted away from the skin, increasing the air layer and providing a greater degree of insulation. In hotter conditions the feathers lie flat against the skin, trapping a smaller amount of air.

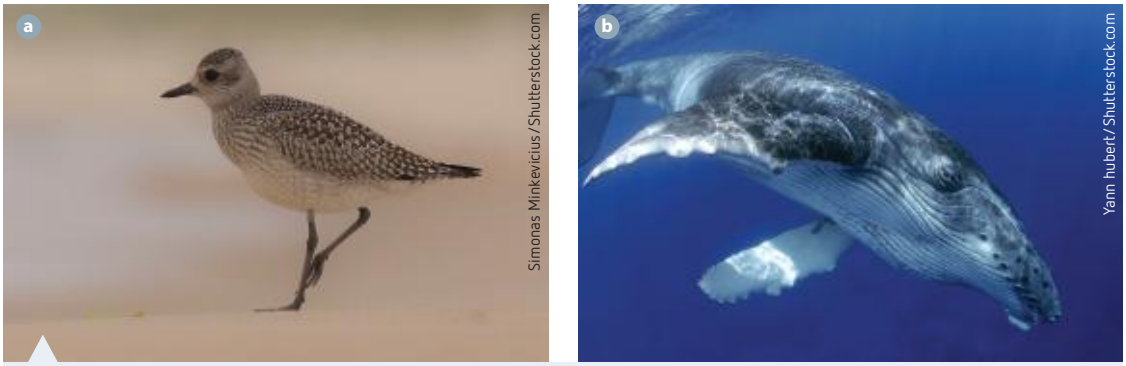


FIGURE 14.29 Endotherms that migrate to avoid severe winter weather conditions include **a** the grey plover and **b** the humpback whale.

Blubber is another form of insulation to reduce heat loss from organisms living in water, such as the Australian fur seal (*Arctocephalus pusillus*) (Fig. 14.30a).

The *surface area to volume ratio* is also an important structural component of temperature regulation.

Animals that live in colder environments, such as the polar bear, are usually larger with a small surface area to volume ratio. This means that there is only a small surface area for heat loss compared to the volume, which allows the body to conserve heat. Smaller animals that live in colder environments, such as the mountain pygmy possum, have small ears to reduce the surface area for heat loss, thereby conserving heat. The mountain pygmy possum (*Burramys parvus*) lives above 1400 metres in the alpine regions of south-eastern Australia. It has short legs, a round body and small ears with limited circulation, which all assist in minimising heat loss (Fig. 14.30b).

Refer to Chapter 3 in *Biology in Focus Year 11* to revise SA:V ratios.

In hotter environments, animals are usually much smaller, with a large surface area to volume ratio, which allows them to lose heat more easily. Many animals that live in hot environments have large, thin ears that allow rapid heat loss. Examples are the bilby (Fig. 14.28) and the African elephant.

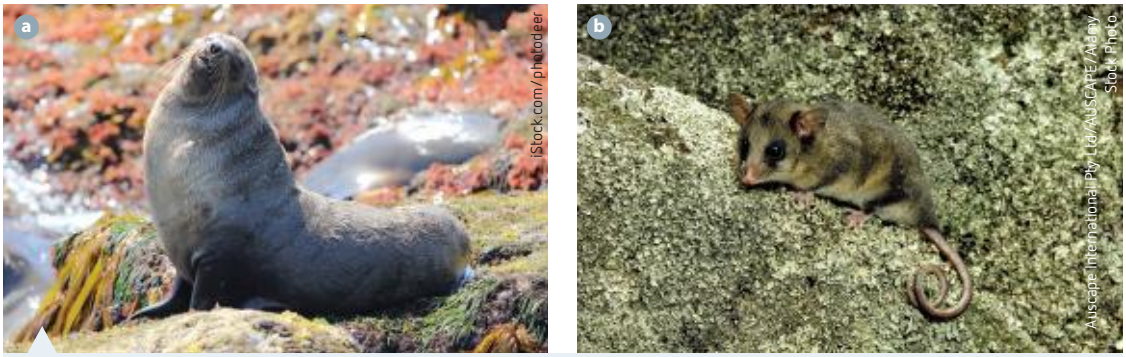


FIGURE 14.30 Structural adaptations to assist in thermoregulation: **a** layers of blubber in the Australian fur seal provide insulation to minimise heat loss; **b** the mountain pygmy possum has a small surface area to volume ratio and small ears, to minimise heat loss.

Physiological adaptations

Physiological adaptations focus on functions within the body. Metabolic activity can be altered to assist the organism in maintaining its body temperature within the tolerance range. In low ambient temperatures, the main source of heat in the body of endotherms is that generated as a result of the metabolic activity

of their cells, particularly the muscle and liver cells. One of the methods by which heat generation occurs is shivering, which, along with increased metabolic rate, produces heat that will increase the body temperature. Conversely, reduced activity by the animal leads to less heat production, which helps to lower the body temperature.

Some organisms use hibernation or torpor to escape the extremes of temperature. **Hibernation** is an extended period of inactivity in response to cold, where the body temperature does not drop below 30°C, but the heart rate and oxygen consumption drop considerably. Hibernation is a mild form of torpor and is less intense, but may last for a longer period of time.

The mountain pygmy possum hibernates during cold winters to escape below-freezing temperatures and to reduce the amount of energy required to keep its body warm.

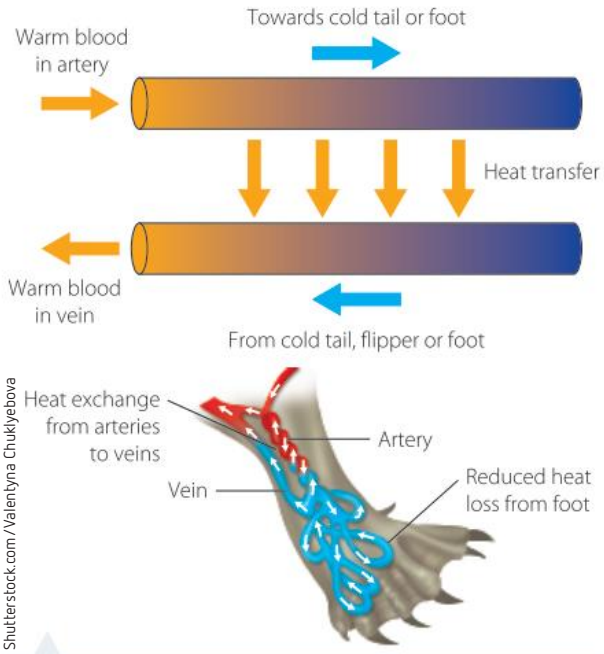
Weblink
Countercurrent exchange

Torpor is a short-term hibernation where the body temperature drops much lower (below 30°C) and metabolism, heart rate and respiratory rate decrease, accompanied by a reduced response to external stimuli.

The common wombat (*Vombatus ursinus*) slows its metabolism to a third of its normal rate on hot days, particularly when sheltering in its burrow. This is a useful strategy, as wombats do not have sweat glands to assist in heat loss. **Evaporative cooling** such as sweating, panting or licking saliva onto the body surface is another common cooling mechanism. As water evaporates from the body, heat is removed.

Organisms can also regulate blood flow to increase or decrease the amount of heat lost to the surroundings. Because blood carries heat, and usually the body temperature of an organism is higher than that of its surroundings, *vasodilation* of capillaries near the skin surface increases the amount of heat released. This mechanism is used in the red kangaroo (along with the behavioural adaptation of licking the forearm to increase heat loss as the saliva evaporates).

Blood flow can also be increased or decreased at extremities to control temperature. The bilby (*Macrotis lagotis*) has an extensive network of capillaries throughout the ear that aid in releasing heat to its surroundings. Furthermore, a mechanism called **countercurrent exchange** (Fig. 14.31) allows the warm blood in arteries (flowing from the heart towards the extremities) to heat the cooler blood in the veins coming back from the cold extremities, before this blood is returned to the heart. This occurs in the feet of the platypus (*Ornithorhynchus anatinus*) as well as the fins of the Australian fur seal, so that the internal core temperature is not lowered by cool blood returning from limbs that have a large surface area exposed to the cold water.



Shutterstock.com/Valentyna Chukyebova

FIGURE 14.31 Countercurrent heat exchanges reduce heat loss at the extremities.

Worksheet
Adaptations to endothermy

KEY CONCEPTS

- Endotherms are organisms that rely on internal sources, such as metabolic activity, to maintain their body temperature within a very narrow range of tolerance limits despite variations in the ambient temperature.
- Adaptations of endotherms to maintain body temperature:

BEHAVIOURAL	STRUCTURAL	PHYSIOLOGICAL
• Change position/ alignment of body • Burrow • Nocturnal activity • Migration	• Insulation • Surface area to volume ratio related to body shape and size of structures such as ears	• Changes to metabolic rate • Hibernation/torpor • Evaporative cooling • Regulation of blood flow to surface and extremities • Countercurrent exchange

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CHECK YOUR UNDERSTANDING 14.3

1 Define the following terms:
   - **a** adaptation
   - **b** endotherm
   - **c** thermoregulation.

2 Unlike endotherms, which are able to regulate their body temperature, the body temperature of *ectotherms* depends on the ambient temperature. On the graph shown in Figure 14.32, identify which line represents each of the following, and explain why:
   - **a** an endotherm
   - **b** an ectotherm.

3 Describe what is meant by the each of following types of adaptations, and provide an example of each in relation to temperature regulation:
   - **a** behavioural
   - **b** structural
   - **c** physiological.

4 Explain why many animals that live in colder environments are much bigger than animals that live in warmer environments.

5 **a** Describe how the process of countercurrent heat exchange works.
   b Explain how this process assists in the maintenance of body temperature. Use a diagram to aid your explanation.

6 The following is a list of adaptations present in a range of endotherms:
   - **i** curls into a ball, limbs drawn in
   - **ii** large, thin ears
   - **iii** burrowing
   - **iv** basking in the sun
   - **v** shivering
   - **vi** panting
   - **vii** red face
   - **viii** lips and nose appear blue
   - **ix** thick fur.

   a Identify which adaptations would be present in animals that live in a cooler environment.
   b For each adaptation you identified in **a**, determine whether it is structural, behavioural or physiological.
   c For each adaptation you identified in **a**, describe how it assists in regulating the body temperature of the endotherm.

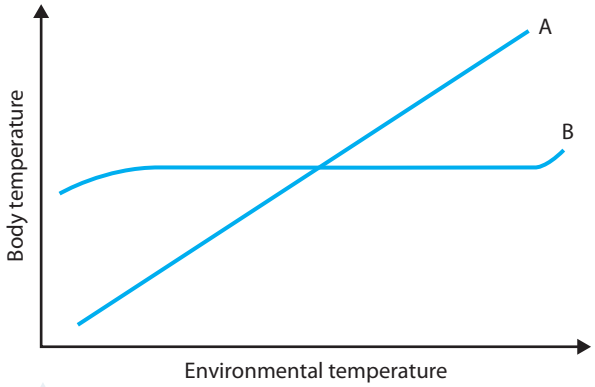

FIGURE 14.32 Body temperature in relation to ambient temperature, for an endotherm and an ectotherm

14.4 Mechanisms to maintain water balance in plants

The main form of water loss in plants is by means of **transpiration** – evaporation of water from the stomata of leaves. Transpiration serves two main functions: it lifts water and dissolved ions from the roots and up the stem to the top of plants in a continuous *transpiration stream*; and it is a form of *evaporative cooling*, a process that is essential in regulating temperature in plants. Stomata also need to be open to allow the exchange of gases for photosynthesis. Plants that live in areas where water is in limited supply (usually hot, dry areas) must achieve a balance between how much water the plant can afford to lose for *cooling* purposes, *transpiration*, *exchange of gases* and the risk of *dehydration*.

Xerophytes are plants that live in arid conditions and have adaptations that equip them to achieve this balance and survive in their hostile environment. Most of these adaptations are evident as modifications of *leaves*, but other organs may also show modifications. Stems and leaf stalks (petioles)

have sparsely distributed stomata, but are green, because they have photosynthetic tissue. This can be used to advantage in allowing xerophytes with reduced leaves to carry out essential functions to survive in their arid habitat.

Reducing internal temperature

Some plants have developed structural features or physiological mechanisms *other than transpiration* to reduce their internal temperature, allowing the plants to use less water for evaporative cooling but still keep their temperature within the correct range for metabolism.

- Their leaves may be coated in a shiny waxy cuticle or a thick, leathery cuticle. This ensures that all the epidermal cells are waterproof, preventing loss of water by evaporation from these surface cells.
- The leaf may have white hairs to reflect sunlight, which will reduce the temperature on the surface of the leaf and thus reduce evaporation.

Reducing the exposure of transpiring plant structures to sunlight

Plant organs that have the most abundant stomata have the greatest rates of transpiration. In some plants, the exposure of these organs (and their stomata) to light is reduced by:

- the orientation of the leaves so that stomata are not exposed to direct light
- reduced surface area of organs that have the highest proportion of stomata
- the complete loss of transpiring plant organs, such as leaves (Fig. 14.33) or leaf-like parts of the plant (for example, flowers). (These plants need to have some additional adaptations to prevent overheating, increase their photosynthetic tissue or ensure pollination, because of their loss of leaf or petal surface area.)

Iee avison/Alamy Stock Photo

FIGURE 14.33 The leaves of some cacti are reduced to spines, to reduce water loss (as well as protecting them against being eaten by animals).

Some examples of adaptations that reduce water loss are:

- *reduced leaf size* – some plants have smaller leaves, with each leaf divided into pinnae or leaflets. Other plants have leaves that are reduced to tiny brown bracts or scales, and their photosynthetic function is taken over by other parts of the plant, such as **cladodes** (photosynthetic stems) and **phyllodes** (photosynthetic leaf stalks). The photosynthetic stems or stalks that take over the function of the leaves have very few stomata and therefore the amount of water lost by transpiration is reduced, while the photosynthetic surface area is still sufficient. Many phyllodes and cladodes have the added features of hairs and/or sunken stomata
- *reduced size of flowers* or *no petals* can reduce the amount of water a plant requires and also reduce evaporation of water from flower surfaces
- *shedding leaves* reduces the overall amount of water lost from leaves
- *orientation of leaves on the stem* helps minimise water loss because the stomata are not directly exposed to sunlight during the hottest part of the day.

Regulating the opening and closing of stomata

Some plants minimise the loss of water by only opening the stomata during the cooler parts of the day, such as early morning and late afternoon. At these times when the temperature is lower, evaporation is reduced and therefore water loss is minimised.

The difference in water concentration between the plant and the surrounding atmosphere determines how much water is lost by transpiration. On a hot, dry day, the water concentration in the air is much lower than in the internal tissues of the leaf, and so more water is lost by transpiration than on a cooler or more humid day.

Because plants cannot change their external environment, many have adaptations that allow them to create their own smaller 'microclimate' in the air immediately surrounding each leaf. Structures such as hairy leaves, rolled leaves and sunken stomata trap water in the immediate vicinity and in this way keep the air around the plant humid, by preventing the moist air being swept away by dry air currents and by creating a barrier to evaporation. These adaptations allow plants to keep their stomata open for a longer period of time, as little water is lost and so gaseous exchange for photosynthesis can occur freely.

Worksheet
Mechanisms to conserve water in plants

Water storage

Some plants, called **succulents**, have adaptations such as fleshy stems or leaves that swell up and retain moisture when it is available; they then survive by using this moisture during dry periods.

Fruits

Fruits are structures that are removed from plants so that the seeds they contain can be dispersed. Many plants produce *woody fruits* rather than fleshy fruits, which reduces the amount of water lost from the plant when the fruits fall off (Fig. 14.34).

FIGURE 14.34 Because the fruit of plants such as banksias is woody and dry, the plant does not lose water when the fruit falls.

INVESTIGATION 14.4

A first-hand and secondary-source investigation into mechanisms in plants to maintain water balance

INTRODUCTION

Many plants possess adaptations that allow water balance to be maintained by minimising water loss while still maintaining necessary functions such as cooling of the plant and photosynthesis.

These adaptations are discussed on pages 497–9.

Information and communication technology capability

Personal and social capability

AIM

To provide examples of adaptations by plants to maintain water balance and explain how these adaptations assist in this process

»

MATERIALS

- samples of leaves with different adaptations for maintaining water balance: hakea, eucalypt, banksia, wattle, casuarina and pigface are some examples that could be used
- binocular microscopes, hand lens
- newspaper
- gloves

RISK ASSESSMENT

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?
Binocular microscope	Sharp edges can cause cuts, damage to limbs if dropped.	Handle with care, ensure glass areas are not chipped or broken.
Plant tissue	Could cause allergic reaction.	Always wear gloves when handling plant tissue. Wash hands immediately after examination.

METHOD

1. Working in groups for this investigation, refer to the information provided on pages 497–9 about the different adaptations of plants to allow water balance to be maintained.
2. Use numerous secondary sources to find specific examples of plants that possess these types of adaptations. Each member of the group should be assigned specific adaptations to research, and the results from all group members can then be collated.
3. You will need to name the plant that possesses the specific adaptation, describe the adaptation, draw a scientific diagram and explain how this adaptation assists in maintaining water balance in the plant.
4. Record your findings in a table like the one supplied in the Results section. This table would be most effective displayed in landscape.

RESULTS

1. Record your results in a table like the one below.

TYPE OF ADAPTATION	NAME OF PLANT	DESCRIPTION OF ADAPTATION	DIAGRAM OF ADAPTATION	HOW THIS ADAPTATION ASSISTS IN MAINTAINING WATER BALANCE
Shiny waxy cuticle on leaf				
Covering of tiny, white hairs on leaf				
Reduced leaf size				
Phyllodes				
Reduced flower size				
Orientation of leaves				
Sunken stomata				
Curled leaves				
Succulent				
Regulation of opening and closing of stomata				

2. Individually observe the specimens of leaves provided at each workstation, and determine the adaptations present on each leaf that would assist in the maintenance of water levels in the plant it originated from.
3. For each leaf, draw a diagram of the adaptation and describe how this adaptation assists in the maintenance of water levels.

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DISCUSSION

1. Discuss the benefits of working as part of a team.
2. Outline the type of environment in which each of the adaptations researched would be necessary for a plant to survive.
3. Plants could conserve water by keeping their stomata closed at all times. Discuss the reasons why the stomata cannot remain closed at all times.

CONCLUSION

Write a few sentences that summarise your findings as related to the aim.

KEY CONCEPTS

- The main form of water loss from plants is transpiration, which is required to move water up the xylem and for evaporative cooling.
- Water is also lost when stomata are open for gas exchange.
- Plants that live in areas of low water availability must achieve a balance between how much water the plant can afford to lose for cooling, transpiration, exchange of gases and the risk of dehydration.
- Xerophytes are plants that live in arid conditions and have adaptations that equip them to achieve this balance and survive in their hostile environment.
- They do this by:
  - reducing the exposure of transpiring plant structures to sunlight
  - reducing the internal temperature
  - regulating the opening and closing of the stomata
  - reducing the difference in water concentration between the plant and the outside air
  - storing water
  - producing woody fruits.

CHECK YOUR UNDERSTANDING 14.4

1. Define the following terms:
   - **a** transpiration
   - **b** transpiration stream
   - **c** xerophytes.
2. Outline the adaptations of plants to achieve the following:
   - **a** reduce the internal temperature
   - **b** reduce the difference in water concentration between the plant and the outside air.

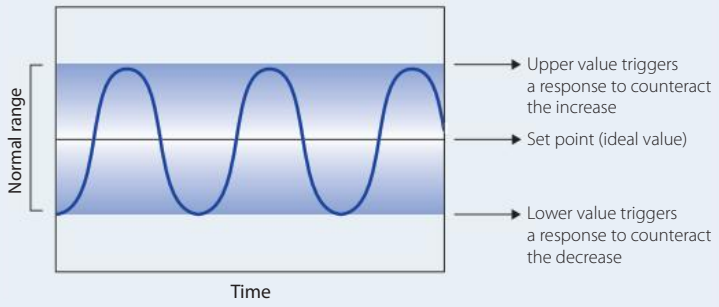
   In each case, explain how these adaptations help to conserve water ,and provide examples of plants that have these adaptations.
3. **a** Distinguish between a *cladode* and a *phyllode*.
   - **b** Explain how cladodes and phyllodes assist the plant to conserve water.
   - **c** Provide examples of plants that possess cladodes or phyllodes.
4. **a** Define 'succulent'.
   - **b** Explain how this type of adaptation assists plants to maintain water balance.

14 CHAPTER SUMMARY

Homeostasis: How is an organism's internal environment maintained in response to a changing external environment?

MAINTAINING HOMEOSTASIS

Homeostasis is the maintenance of a relatively constant internal state, regardless of external changes.

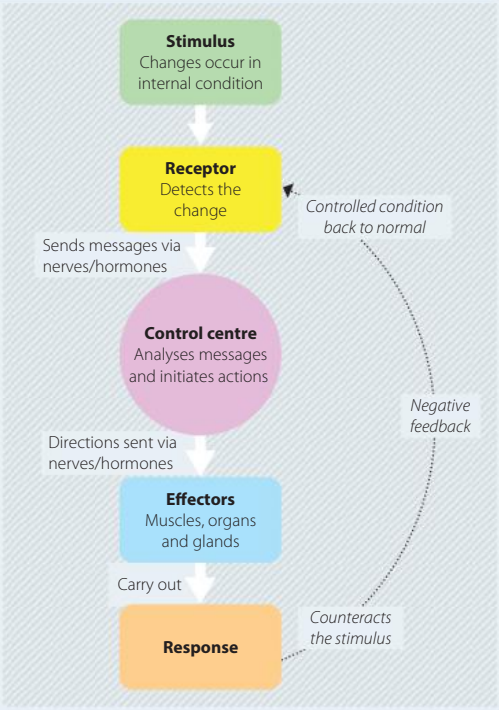


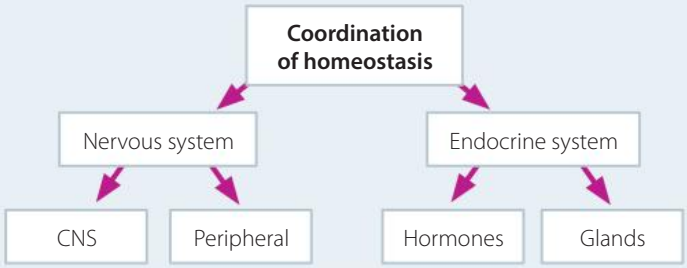
It is important to maintain internal conditions at a level that allows optimal functioning of enzymes to ensure optimal metabolic efficiency.

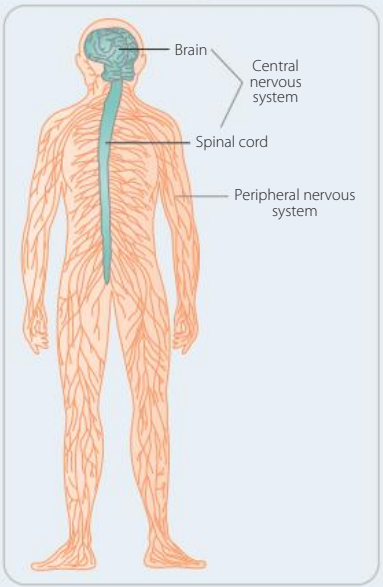
The two main stages of homeostasis are:

- detecting change
- counteracting change.

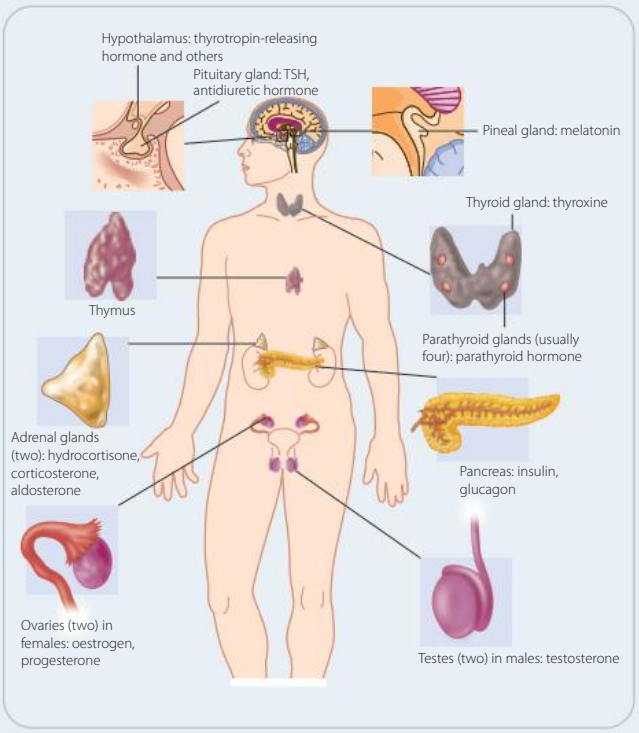
The body maintains homeostasis using negative feedback mechanisms, e.g. maintenance of body temperature and glucose level.







COORDINATION BY THE ENDOCRINE SYSTEM



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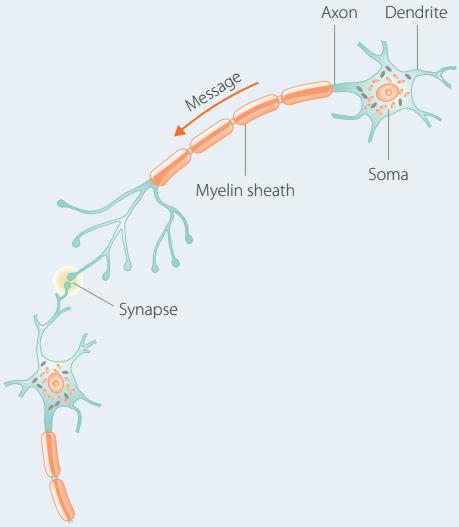
COORDINATION BY THE NERVOUS SYSTEM

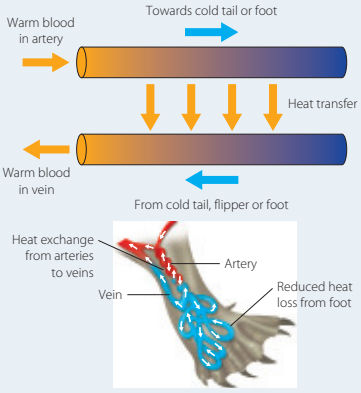


- **Neurons** are nerve cells that make up the nervous system and carry electrical messages in the form of electrical impulses.
- A typical neuron consists of a cell body, dendrites and axon.
- There are three main types: sensory, motor, interneurons.
- A synapse is the gap between the axon of one neuron and the dendrites of a neighbouring neuron.
- Neurotransmitters transfer impulses across the synapses.

MAINTAINING BODY TEMPERATURE

Adaptations to maintain body temperature – endotherms

BEHAVIOURAL	STRUCTURAL	PHYSIOLOGICAL
• Change position/ alignment of body • Burrow • Nocturnal activity • Migration	• Insulation • Surface area to volume ratio related to body shape and size of structures such as ears	• Changes to metabolic rate • Hibernation/torpor • Evaporative cooling • Regulation of blood flow to surface and extremities • Countercurrent exchange



MAINTAINING WATER BALANCE

Mechanisms to maintain water balance in plants

Need to find a balance between transpiration, exchange of gases, evaporative cooling and the need to conserve water

- Reducing internal temperature
- Reducing exposure to sunlight
- Regulating the opening and closing of stomates
- Water storage
- Fruits

14 CHAPTER REVIEW QUESTIONS

Review quiz

1 Describe the importance of homeostasis in living organisms.

2 Explain, using an example, what is meant by a negative feedback mechanism and its importance in living systems.

3 Explain the relationship between metabolic rate and temperature regulation in birds and mammals.

4 Assess the importance of the role of the nervous system in maintaining homeostasis.

5 **a** Using the information provided on page 491 about the secretion of aldosterone to maintain the balance of sodium (Na^+) and potassium (K^+) ions in the blood and your own research, construct a negative feedback loop to summarise the steps that occur:
   - **i** when levels of Na^+ are too high
   - **ii** when levels of Na^+ are too low.

   Hint: You may wish to include both loops in the one diagram, like the one shown for temperature regulation in Figure 14.4.

   b For each of the loops you have drawn for question **a**, identify the:
   - **i** stimulus
   - **ii** receptor
   - **iii** control centre
   - **iv** effector
   - **v** response.

6 Discuss how the endocrine system assists in maintaining the health and wellbeing of organisms.

7 At times, endocrine glands malfunction, causing secretion of incorrect levels of hormones. Research the effects on the body of the following:
   - **a** The thyroid gland secretes:
     - **i** too much thyroxine
     - **ii** too little thyroxine.
   - **b** Too much cortisol is circulating in the bloodstream.
   - **c** The adrenal cortex does not produce enough aldosterone.

8 **a** Research and identify the types of adaptations possessed by endotherms that help them to balance their water levels in hot, dry environments.
   - **b** Explain how each of the adaptations you identified in **a** assists in maintaining water balance.
   - **c** Support your research with named examples and the adaptations they possess. Include at least one behavioural, structural and physiological adaptation.

9 Assess the importance of adaptations by an organism in helping to regulate internal body conditions such as temperature.

10 Research information about *sclerophyll* plants and answer the following questions.
   - **a** What is a sclerophyll plant?
   - **b** Provide two Australian examples of sclerophyll plants and identify some adaptations that each of these plants possesses to assist in temperature regulation.
   - **c** Explain how each of these adaptations assists the plant to conserve water.

11 Discuss the processes that a plant has to balance in order to maintain proper functioning while conserving water.

12 Refer to the graph in Figure 14.35.

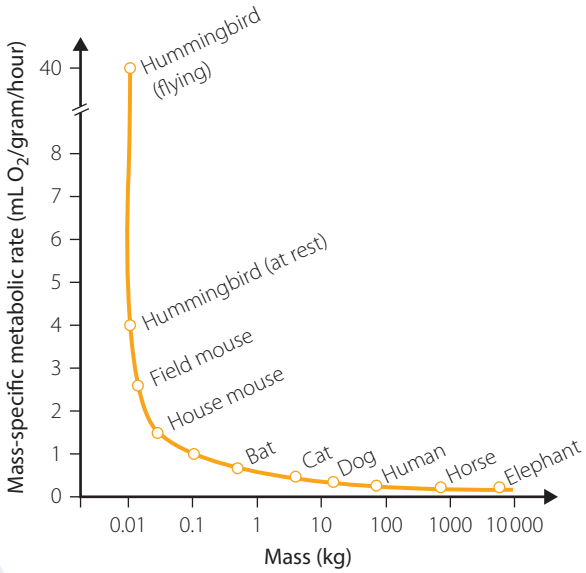


FIGURE 14.35 Graph comparing mass of an organism with metabolic rate

   - **a** Make a general statement about the trend shown in this graph.
   - **b** Explain why an elephant has a lower metabolic rate than a house mouse.

13 Refer to the diagrams in Figure 14.36.

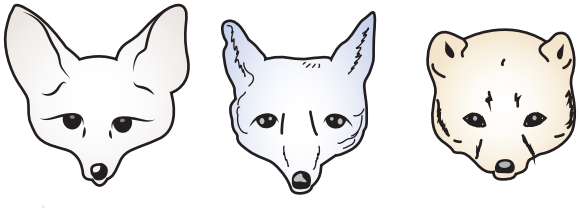


FIGURE 14.36 Organisms in different habitats exhibit different structural adaptations.

   - **a** Identify which of the organisms would most likely live:
     - **i** in a hot, dry environment
     - **ii** in a cold environment.
   - **b** Justify each of your choices in **a**.

Exam preparation

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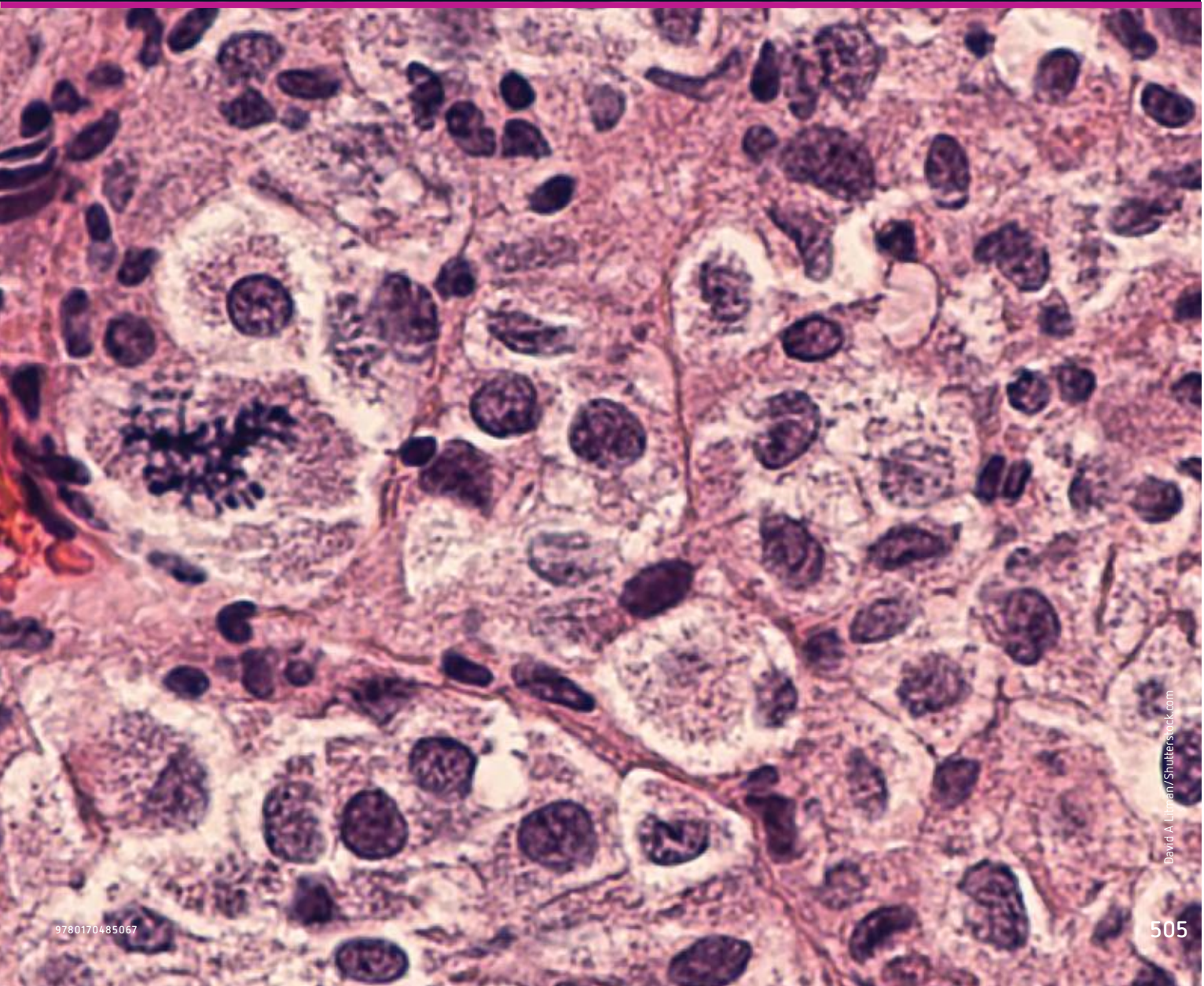
15 Non-infectious diseases

INQUIRY QUESTION

Do non-infectious diseases cause more deaths than infectious diseases?

Students:

- investigate the causes and effects of non-infectious diseases in humans, including but not limited to: ICT
  - genetic diseases
  - diseases caused by environmental exposure
  - nutritional diseases
  - cancer
- collect and represent data to show the incidence, prevalence and mortality rates of non-infectious disease, for example: ICT, N, CCT, DD
  - nutritional diseases
  - diseases caused by environmental exposure



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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
15.1 A secondary-source investigation into the causes and effects on the body of non-infectious disease

15.2 A secondary-source investigation to analyse data on the incidence, mortality and prevalence of melanoma

15.3 A secondary-source investigation into the incidence, prevalence and mortality rates of nutritional diseases

15.4 A secondary-source investigation to determine whether non-infectious diseases cause more deaths than infectious diseases

Worksheets
- Diseases with a genetic cause
- Non-infectious diseases caused by environmental exposure
- Diseases caused by nutritional deficiency
- The role of genes in the formation of cancer

Nelson MindTap

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Non-infectious diseases account for nearly two-thirds of deaths world-wide. In developed countries these diseases stem from a shortlist of 21st century risk factors such as poor diet, tobacco use, alcohol consumption and physical inactivity. In developing countries, the risk factors for disease (both infectious and non-infectious) are the same as those that have dogged humanity through the centuries: lack of food, limited access to clean water (Figure 15.1) and poor hygiene. By studying the incidence, prevalence and mortality rates of disease, we can better understand where and how to intervene to make the most difference.

FIGURE 15.1 Lack of access to clean water is a serious risk factor in developing countries.

15.1 Causes and effects

Non-infectious diseases (also known as non-communicable diseases) are not caused by a pathogen and are not contagious. There is a huge range of non-infectious diseases, and their causes are many and varied. Table 15.1 shows that the top four leading causes of death in Australia in 2015 were all non-infectious

TABLE 15.1 Leading causes of death in Australia in 2015

CAUSE OF DEATH	NUMBER OF PERSONS	PROPORTION OF TOTAL DEATHS (%)
All causes	*159 052*	*100*
Ischaemic heart disease	19 777	12.4
Dementia, including Alzheimer's disease	12 625	7.9
Cerebrovascular disease (stroke)	10 869	6.8
Trachea, bronchus and lung cancer	8 466	5.3
Chronic lower respiratory diseases (including asthma and emphysema)	7 991	5.0
Diabetes	4 662	2.9
Colon, sigmoid, rectum and anus cancer	4 433	2.8
Blood and lymph cancer (including leukaemia)	4 412	2.8
Heart failure	3 541	2.2
Diseases of the urinary system	3 433	2.1
Prostate cancer	3 195	2.0
Influenza and pneumonia	3 042	1.9

Source: Australian Bureau of Statistics (3303.0) 'Australia's leading causes of death, 2015', Table 2.1, 'Leading causes of death'. ©Comonwealth of Australia (CC BY 2.5 Australia)

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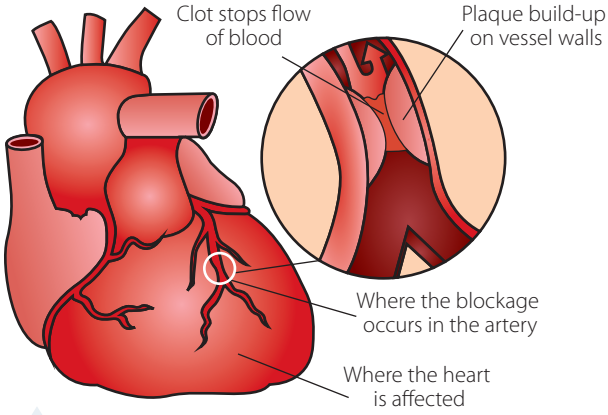
diseases. The greatest proportion of deaths was caused by ischaemic heart disease, with the next most common being dementia.

The risk of developing a specific non-infectious disease is influenced by factors such as age, gender, economic and social conditions, culture, race, lifestyle, environment, genetics and nutrition.

Many non-infectious diseases are considered to be preventable as they are caused by factors that can be modified. For example, reduced exposure to UV rays will reduce the chance of developing skin cancer. The risk of developing diabetes mellitus type 2 (or diabetes type 2) can be reduced by maintaining a normal weight, doing regular exercise and having a well-balanced diet.

Non-infectious diseases are caused by a multitude of factors. Genetic diseases, nutritional diseases, diseases caused by environmental exposure, including lifestyle factors, and cancer are some common types of non-infectious diseases with varying effects on the individual. These are discussed below.



Maniki_rus/Shutterstock.com

FIGURE 15.2 Ischaemic heart disease was the major cause of death in Australia in 2015.

A number of other types of non-infectious disease are not covered in this chapter. These include diseases that cause some form of physiological malfunction, such as when glands malfunction and cause a hormonal imbalance, which would interfere with the maintenance of homeostasis in the body. Diseases such as mental illnesses and those associated with the process of ageing, such as dementia, are also non-infectious.

Weblink
Non-infectious diseases
Compare non-infectious disease in different countries of the world, the risk factors and changes in disability and death caused by non-infectious disease.

Genetic diseases

Genetic diseases are caused by mutations of the genes or chromosomes of an individual. Each gene codes for the production of a specific protein, which may be structural, an enzyme or a hormone. If the particular gene that codes for a specific protein is in some way abnormal then that protein may not be produced correctly and genetic disease may result.

Genetic disease may also result when cell division, by mitosis or meiosis, occurs abnormally, resulting in cells with an incorrect number of chromosomes or abnormal chromosomes.

Refer to mutations of genes and chromosomes in Chapter 7.

Single-gene abnormalities

Many single-gene genetic diseases in individuals are caused when the person inherits mutated genes from their parents. Some of these diseases are caused by abnormal recessive alleles, so the disease will only be expressed if the individual receives the same abnormal recessive allele from each parent. If the abnormal gene is dominant then the individual only needs to receive the abnormal allele from one parent for the disease to be expressed.

Weblink
Genetic diseases
Investigate germline and somatic cell mutations causing genetic disease.

If the abnormal gene occurs on one of the sex chromosomes (normally the X chromosome), the disease is termed 'sex-linked'.

Many metabolic processes that occur in the body are made up of a series of reactions that follow on from each other. Enzymes, the production of which is coded for by genes, catalyse each of these single reactions. If one of the enzymes in the pathway is coded for by a mutated gene, then that enzyme will not be manufactured correctly and the metabolic pathway may not proceed as it should, resulting in genetic disease.

Refer to information on inheritance patterns in Chapter 5.

Diseases caused by gene mutations include cystic fibrosis, albinism, Huntington's chorea, thalassaemia, Tay Sachs disease, sickle cell anaemia, phenylketonuria and the sex-linked diseases Duchenne muscular dystrophy and haemophilia.

Cystic fibrosis is an inherited disease caused by mutation of the cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7. This faulty gene changes the protein that regulates the normal movement of sodium chloride in and out of cells. This causes the mucus-secreting

organs to produce abnormally thick mucus. The CFTR gene is recessive, meaning that a person must have two copies of the faulty gene for them to develop the disease.

Cystic fibrosis affects the respiratory, digestive and reproductive systems, as well as the sweat glands. The mucus secreted is very thick and blocks passageways in the lungs and digestive tracts (Fig. 15.3). This thick mucus causes problems with breathing and with the digestion and absorption of nutrients.

Some of the symptoms of cystic fibrosis are:

- recurrent and severe chest infections
- malabsorption of nutrients and failure to thrive
- passing of large, foul-smelling motions
- diabetes
- excess salt in sweat
- liver failure
- infertility.

The average life expectancy of someone suffering from cystic fibrosis is 30–40 years.

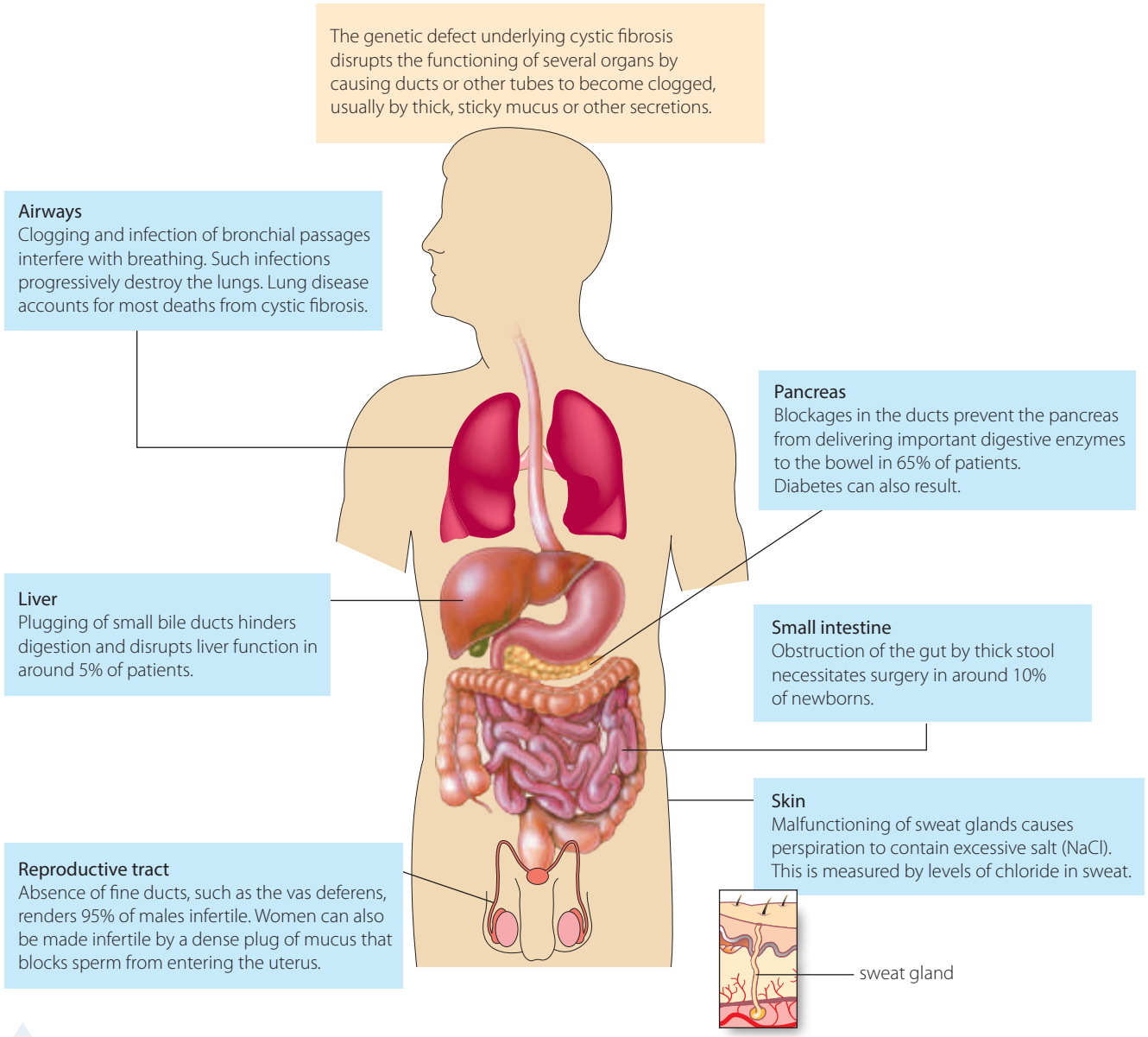


FIGURE 15.3 Organs affected by cystic fibrosis

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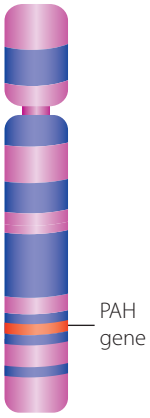
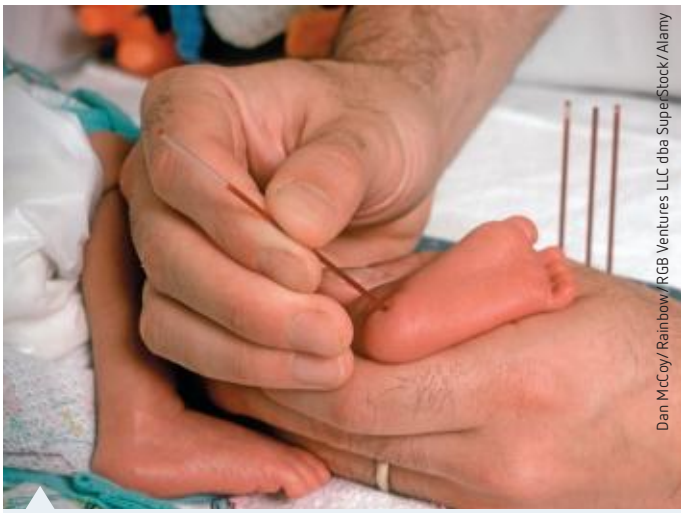
Phenylketonuria (PKU) is a genetic disease caused by a mutation of the gene on chromosome 12 that codes for an enzyme called **phenylalanine hydroxylase** (PAH) (Fig. 15.4). This enzyme is required for the first step in the breakdown of the amino acid **phenylalanine** (a component of some proteins). As a result of the defective gene, the PAH enzyme is not manufactured correctly and the phenylalanine cannot be broken down. This leads to a dangerous build-up of phenylalanine in the blood and tissues, resulting in, among other things, the development of intellectual disabilities, delays in development, social, emotional and behavioural problems, psychiatric disorders, hyperactivity and the risk of seizures. PKU is an autosomal recessive disease and all newborns are tested for this condition (Fig. 15.5). If an individual is diagnosed with PKU they must follow a diet low in protein-rich foods such as milk, nuts, eggs and meat for the rest of their lives. This prevents a build-up of phenylalanine. The person must also avoid using sugar-free products that contain artificial sweeteners made with phenylalanine.

Weblink
Understanding PKU
Click your way through the presentation and summarise the information presented about PKU in a table.



Chromosome 12

FIGURE 15.4 Phenylketonuria is caused when a defective PAH gene on chromosome 12 is inherited.



Dan McCoy/Rainbow/RGB Ventures LLC dba SuperStock/Alamy

FIGURE 15.5 All newborns are tested for PKU by taking a blood sample from the heel.

Chromosomal abnormalities

Some genetic diseases are caused by mutations of the chromosomes that carry the genes. These mutations can take the form of:

- an incorrect number of chromosomes – too many or not enough
- a change to the chromosome itself – a deletion, addition or altered sections.

An incorrect number of chromosomes may come about due to:

- **non-disjunction** – the incorrect separation of chromosomes during cell division. The end result of this process is an incorrect number of chromosomes in the zygote
- **trisomy** – when there is one extra chromosome along with the normal chromosome pair and the total number of chromosomes present in the zygote is one more than the normal diploid number of chromosomes
- **monosomy** – when one member of a chromosome pair is missing and the total number of chromosomes in the zygote is one less than the normal diploid number.

Chromosome mutations are covered on pages 238–40.

Worksheet
Diseases with a genetic cause

Many embryos formed with an incorrect number of chromosomes will spontaneously miscarry. Embryos with monosomy or trisomy that are carried to full term will have a genetic disease or syndrome (Table 15.2). A **syndrome** is a group of symptoms that occur together and characterise a particular disease. The chromosome number that forms the trisomy or monosomy determines the type of syndrome.

TABLE 15.2 Trisomy and monosomy syndromes and the chromosome involved in each

SYNDROME	CAUSE
Patau	Trisomy 13
Edwards	Trisomy 18
Down	Trisomy 21
Triple X	Trisomy X (XXX)
Klinefelter	XXY
Jacob	XYY
Turner	Monosomy X (XO)

Each of these syndromes is characterised by a different set of symptoms. Turner syndrome is the only full monosomy syndrome seen in humans. All other full monosomy syndromes are lethal and cause spontaneous miscarriage.

Down syndrome is the most common trisomy syndrome (1 in 800 live births). It is caused by the presence in the zygote of an extra chromosome 21, or part of this chromosome, meaning that there are three copies of chromosome 21 in every cell, instead of the normal two copies (Fig. 15.6a). It is named after the first person to recognise and describe the condition, Langdon Down, in 1866.

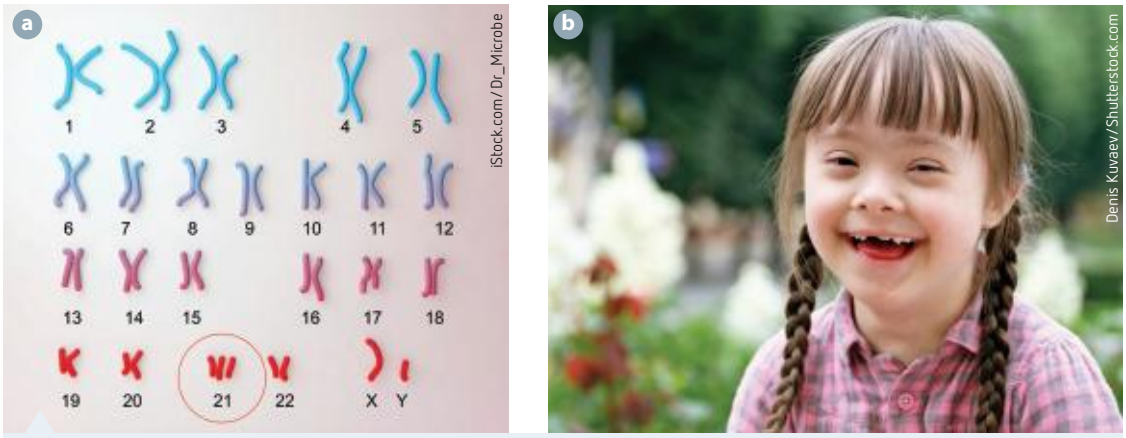


FIGURE 15.6 Down syndrome is a genetic disease caused by an incorrect number of chromosomes, trisomy 21. **a** The karyotype of a person with Down syndrome showing an extra chromosome 21. **b** Characteristic facial features of a person with Down syndrome

The effects of Down syndrome vary in individuals but include many of the following:

- characteristic facial features including a small flattened skull, a short flat-bridged nose, wide set, almond-shaped eyes and skin folds on the eyes (Fig. 15.6b)
- a protruding tongue
- small ears that fold over a bit at the top

- intellectual disability
- short stature
- heart defects
- sexual underdevelopment and infertility
- short, stubby hands
- looseness in joints
- susceptibility to infection.

Klinefelter syndrome is a trisomy involving sex chromosomes. Males normally have two sex chromosomes – one X and one Y (XY). Males with Klinefelter syndrome have three sex chromosomes – two X and one Y (XXY) (Fig. 15.7). These males have lower levels of testosterone and unusually small testes, and are sterile. They are taller than average height, have some degree of breast enlargement and feminine fat distribution on the abdomen and hips. Young boys with Klinefelter often have behavioural and learning problems.

Individual chromosomes can also be changed or mutated. Sometimes the chromosome breaks and some of it is deleted. When this happens, the genes that are present on the deleted section have been lost. Such a chromosome is said to have a **deletion**.

Cri du chat ('cry of the cat') syndrome is a rare disease that results from the deletion of a specific section of chromosome 5. Individuals with this deletion have intellectual disabilities, a small head, flattened bridge of the nose, wide-spaced eyes, receding chin and a cry that sounds like a distressed cat.

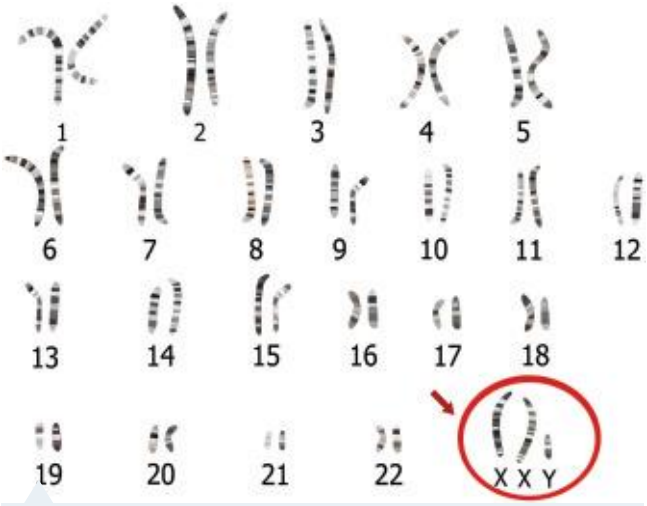


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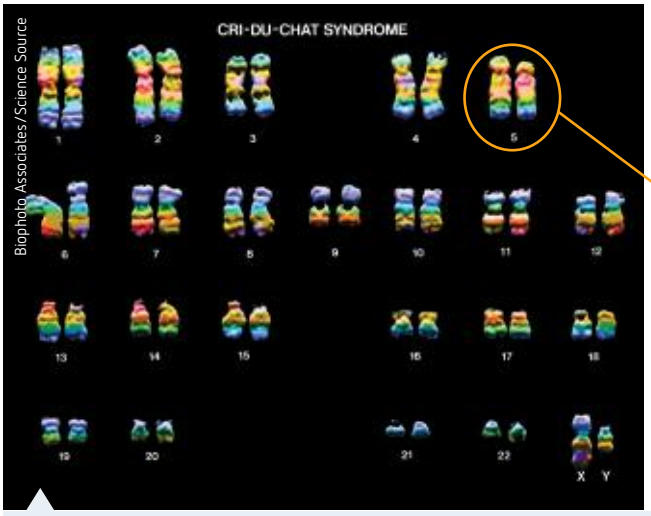
FIGURE 15.7 Karyotype of Klinefelter syndrome, a trisomy involving an extra X chromosome



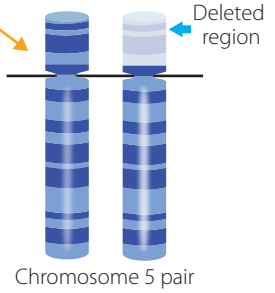


FIGURE 15.8 Cri du chat syndrome: **a** karyotype; **b** the disease is caused by deletion of part of chromosome 5.

Chromosomes can also be mutated when a section of one chromosome moves to join onto another chromosome in a different homologous pair. This process is called **translocation**. No genes are lost in this process but the genes on the section of chromosome that has translocated are now combined with different genes, which could alter the phenotype expressed.

For example, a translocation of the bottom portion of chromosome 9 to the bottom of chromosome 22 causes acute myelogenous leukaemia.

Weblink
Genetic disorders
Use information to describe different genetic disorders.

KEY CONCEPTS

- Non-infectious (non-communicable) diseases are not caused by a pathogen and are not contagious.
- Non-infectious diseases are the leading cause of deaths globally
- Risk factors for developing a specific non-infectious disease are age, gender, economic and social conditions, culture, race, lifestyle, environment, genetics and nutrition.
- Genetic diseases are caused by mutations to the genes or chromosomes of an individual.
- Mutated genes may be inherited and cause genetic disease due to incorrect proteins (or no proteins) being formed, interrupting metabolic reactions. Examples are cystic fibrosis and phenylketonuria.
- Chromosomal abnormalities include an incorrect number of chromosomes, or additions or deletions of chromosomes.
- In trisomy, the individual has one more chromosome than required; in monosomy, there is one less chromosome than required.
- Down syndrome is caused by an extra chromosome 21, Klinefelter syndrome by the presence of three sex chromosomes (XXY) instead of the usual two, and Cri du chat syndrome by deletion of a section of chromosome 5.

CHECK YOUR UNDERSTANDING 15.1a

1. Define 'non-infectious disease'.
2. Identify the three leading causes of death in Australia in 2015.
3. List some risk factors associated with the development of non-infectious disease.
4. a. Define 'genetic disease'.
   b. Outline the two main ways in which genetic disease is caused.
5. Describe how a mutation to a single gene is capable of causing a disease.
6. Outline the cause and effect on the body of the genetic disease:
   a. cystic fibrosis
   b. Klinefelter syndrome.

Diseases caused by environmental exposure

Humans interact with their environment throughout their lives. Numerous factors that individuals are exposed to in the environment surrounding them can cause diseases of different types.

There are many different types of environmental diseases, including:

- lifestyle diseases, such as cardiovascular disease and diseases caused by substance abuse such as alcoholism and smoking-related diseases
- diseases caused by physical factors in the environment, such as skin cancer caused by excessive exposure to ultraviolet radiation in sunlight
- diseases caused by exposure to chemicals in the environment, home or workplace, such as lead poisoning caused by exposure to high levels of lead in the atmosphere, soil and products used in everyday life.

Lifestyle diseases

Lifestyle diseases are those that arise as a direct result of the way in which individuals live their lives. The most common lifestyle diseases are cardiovascular disease, diabetes, cancers and chronic lung disease. Risk factors that lead to the development of these diseases are tobacco use, an unhealthy diet, physical inactivity and the harmful use of alcohol. Exposure to additives and hormones added to our food products may also have a detrimental effect.

One type of cardiovascular disease is **atherosclerosis**, commonly known as 'hardening of the arteries'. This disease can be caused by a variety of factors in a person's lifestyle, including insufficient physical

activity, drinking alcohol excessively, being exposed to high levels of stress, smoking, and having an unbalanced diet that could be too high in kilojoules, saturated fats, salt or red meat.

Following these lifestyle activities for a prolonged period of time leads to the deposition of lipids (fatty deposits) in the inner walls of arteries. With continued deposition, the internal walls of the arteries are no longer smooth but very rough and thickened, which hinders blood flow and increases blood pressure (**hypertension**). **Plaque**, a hard, calcified substance, is deposited on the artery walls and further reduces elasticity and blood flow (Fig. 15.9). This further increases hypertension, which can cause **cerebral haemorrhage**, the leakage of blood into brain tissue. Blood clots are also prone to form and, along with the build-up of fat and plaque, can cause a blockage (**occlusion**) in the blood vessel. This occlusion could be a partial blockage or a total blockage, causing numerous problems for the functioning of the body. If the blockage occurs in the arteries supplying the heart with blood (**coronary** arteries), this may result in **cardiac arrest** or heart failure. If the occlusion occurs in the arteries in the brain, this may result in a stroke. Atherosclerosis of the coronary arteries is called **ischaemic heart disease**, while atherosclerosis of the cerebral arteries is called **cerebrovascular disease**.

Worksheet
Non-infectious diseases caused by environmental exposure

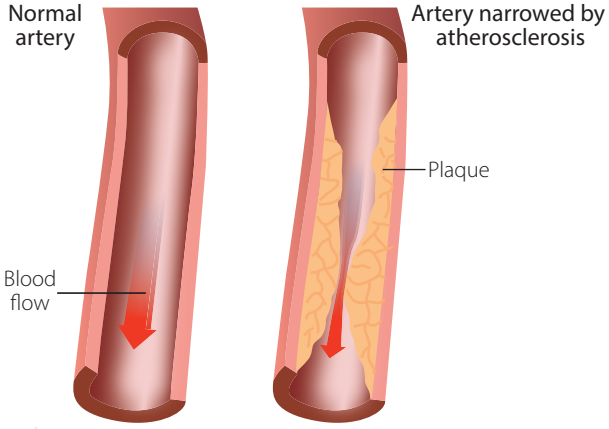


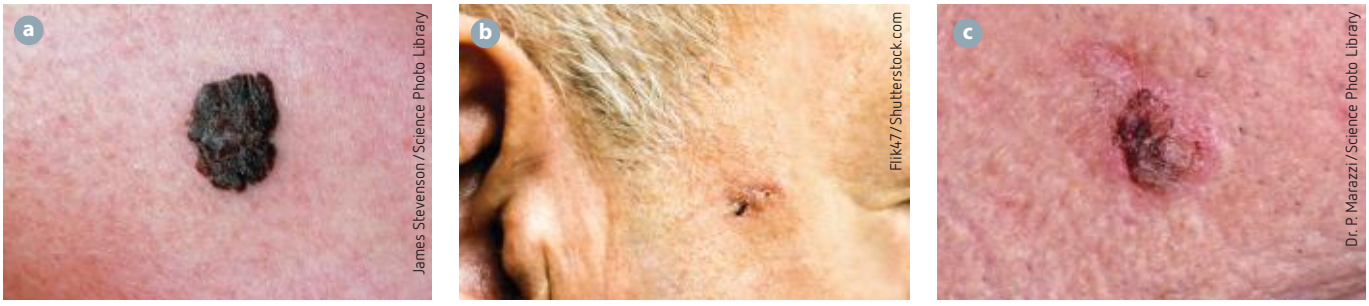
FIGURE 15.9 Atherosclerosis involves the build-up of plaque on the internal surfaces of blood vessels, restricting blood flow.

Physical factors

Exposure to physical factors in the environment can cause disease. Some of these physical factors include ultraviolet (UV) light from the sun or in solariums, or radiation from nuclear substances. These physical factors often affect the health of the individual by changing the genetic material in some way, causing problems with the correct functioning of the body.

Excessive exposure to UV light can cause skin cancer (Fig. 15.10). The UV light causes changes to the DNA of the skin cells, which may cause continued abnormal cell division. This can lead to the formation of basal cell carcinomas, squamous cell carcinomas or malignant melanomas. The most serious of these is malignant melanoma, which can spread to other areas of the body unless identified and treated early.

Weblink
Atherosclerosis
Watch the video to summarise the processes involved in the formation of atherosclerosis.



Melanoma | Basal cell carcinoma | Squamous cell carcinoma

FIGURE 15.10 The most common types of skin cancer: **a** melanoma, **b** basal cell carcinoma and **c** squamous cell carcinoma

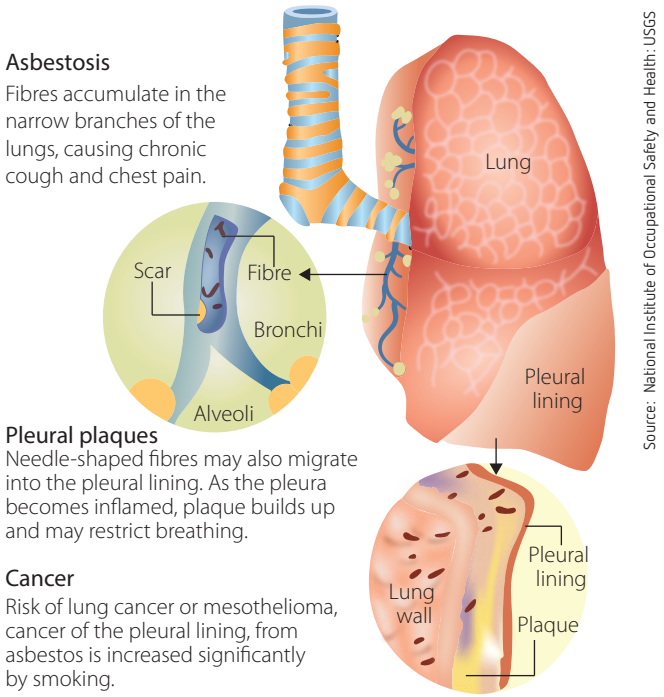
Exposure to chemicals

The environment around many of us contains a vast array of chemicals. Exposure to some of these chemicals can cause disease – the type of disease and the period of exposure necessary will vary depending on the chemical. Many factors in the environment can be the cause of disease – it might be that the land that homes are built on has previously been polluted unwittingly or has been used to dispose of waste that has then been buried. It might be toxic discharges into the atmosphere or waterways as a

result of industrial processes, or exposure to chemicals in the workplace. Pollution of the air and water causes many diseases worldwide, more so in countries where pollution is not regulated.

Asbestos is a chemical that historically was present in many products in common use. It is now known that exposure to even a small amount of asbestos may cause one of the asbestos-related diseases: asbestosis, lung cancer or **mesothelioma** (a cancer that occurs in the membrane surrounding the lungs) (Fig. 15.11). The chance of developing these diseases increases with prolonged exposure to asbestos. The effects of inhaling asbestos fibres does not become obvious until many years later.

FIGURE 15.11
Damage to the lungs caused by asbestos



Weblink
Toxtown
Determine the chemicals present in the different environments of Toxtown

Asbestosis is caused when inhaled asbestos fibres cause an inflammatory reaction in the lung tissue. This inflammation leads to scarring and stiffening of the lung tissue, which makes the process of breathing much harder, and reduces the amount of oxygen taken up into the blood. This causes noticeable symptoms such as shortness of breath, a persistent cough and chest pain, along with appetite loss and enlarged fingertips (finger clubbing) caused by a lack of oxygen to the extremities.

Asbestosis can lead to the development of mesothelioma.

Prolonged exposure to heavy metals in the environment can also cause disease. Heavy metals such as lead and mercury occur naturally on Earth and are used in many manufactured products. Many substances that the body does not need can be excreted. However, these heavy metals cannot be easily excreted and will build up in the body with prolonged exposure.

Historically, lead has been a common ingredient in paint and fuel. Although that is not still the case, lead is still used in batteries, solder, some roofing material, pipes, as a glaze on pottery and in some imported toys.

Children can be exposed to lead in the soil, water, old paint, pottery and some toys. Lead builds up to dangerous levels in the body before there are any noticeable symptoms.

Toxic levels of lead in the body (Fig. 15.12) can cause:

- developmental delays
- learning difficulties
- irritability

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- fatigue and sluggishness
- weight loss and loss of appetite
- abdominal pain, vomiting and constipation.

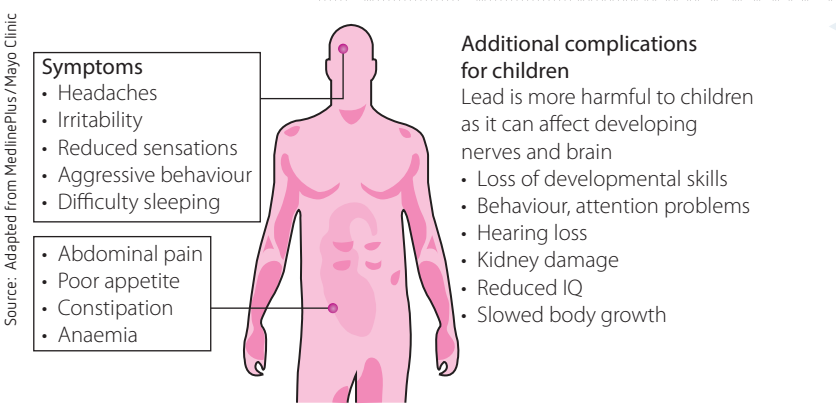


FIGURE 15.12 The effects of lead poisoning

Weblink
Lead poisoning
Watch the video and summarise the effects of lead poisoning

KEY CONCEPTS

- Diseases caused by environmental exposure include lifestyle diseases and diseases caused by exposure to both physical and chemical factors in the environment.
- Lifestyle diseases are caused by how individuals live their lives, and include cardiovascular disease, diabetes, cancers and chronic lung disease.
- Risk factors that lead to the development of these diseases are tobacco use, an unhealthy diet, physical inactivity and the harmful use of alcohol.
- The lifestyle disease atherosclerosis is caused by a variety of factors including insufficient physical activity, drinking excessively, being exposed to high levels of stress, smoking and consuming an unbalanced diet (too high in kilojoules, saturated fats, salt or red meat).
- Exposure to physical factors in the environment, such as UV rays and nuclear radiation, can cause disease by changing the genetic material (e.g. exposure to UV light can cause skin cancer).
- Exposure to chemicals in the environment can cause diseases such as mesothelioma, asbestosis and lead poisoning – the type of disease and the period of exposure necessary will vary depending on the chemical.

CHECK YOUR UNDERSTANDING 15.1b

1 Identify and outline the main types of diseases caused by environmental exposure.
2 Describe the cause and effect on the body of an example of a:
   a lifestyle disease
   b disease caused by physical factors in the environment
   c disease caused by exposure to chemicals in the environment.
3 Explain why lifestyle diseases are said to be preventable.

Nutritional diseases

To maintain health and wellbeing, it is essential to consume the correct balance of nutrients. Eating a balanced diet that contains the recommended amount and type of food provides all the nutrients required for all necessary bodily processes (Fig. 15.13).

Proper nutrition contributes to better infant, child and maternal health. It also contributes to a stronger immune system and a reduced risk of developing a non-infectious disease, such as diabetes or cardiovascular disease.

Nutritional diseases are caused by diets lacking the proper balance and amount of nutrients. They can also be caused by psychological conditions that lead to inappropriate diets. This imbalance in the diet leads to a condition known as **malnutrition**.

There are two broad categories of malnutrition:

- undernutrition – insufficient intake of the correct type of food, quantity of food, or a combination of both
- overnutrition – excessive intake of food.

FIGURE 15.13 The Australian Guide to Healthy Eating provides a visual representation of the recommendations for the proportions of the five food groups to be consumed each day.

Undernutrition

There are a number of types of undernutrition, described below.

FIGURE 15.14 This child is suffering from the effects of kwashiorkor, a disease caused by insufficient protein in the diet

Lack of protein and energy-rich food

In developing countries, where food is often in short supply for numerous reasons, many individuals, especially children, suffer from a disease known as **kwashiorkor**. This is caused by a severe lack of protein in the diet and results in a failure to grow, enlarged liver, hair changes, apathy, irritability and increased susceptibility to infectious diseases. Kwashiorkor is characterised by a swollen belly, caused by fluid retention (**oedema**).

Gross food deprivation leads to a severe lack of both protein and energy intake. This causes a disease called **marasmus**, in which the sufferers are extremely underweight and have lost most of their subcutaneous fat. They are very weak and susceptible to infection, and if untreated this will lead to death by starvation or heart attack.

Kwashiorkor and marasmus can also occur in patients recovering in hospital and receiving glucose intravenously for an extended period of time, or those suffering from diseases that suppress the appetite or cause malabsorption of nutrients.

Lack of vitamins

A diet that is lacking a vital nutrient, usually a vitamin or a mineral, can lead to a nutritional deficiency disease. The particular vitamin or mineral that is lacking will determine the disease or condition that occurs in the individual. Some examples of vitamin deficiencies are outlined below.

Vitamin A is important for normal growth, healthy skin and mucous membranes, and normal vision, including night vision. A lack of vitamin A in the diet can lead to blindness, dry skin and increased susceptibility to infection. It is a common cause of blindness in children in developing countries.

Vitamin D is produced by the body in a series of reactions initiated by exposure to the sun. With limited exposure to the sun and a diet lacking in vitamin D, children can develop a disease called **rickets** that results in defective calcification of bones, retardation of growth and deformities of parts of the skeleton, such as bowed legs (Fig. 15.15). In adults, a lack of vitamin D causes a disease known as **osteomalacia**, which not only causes problems with the bones but also weakens muscles. **Osteoporosis**, thinning of the bones, has some association with inadequate vitamin D.

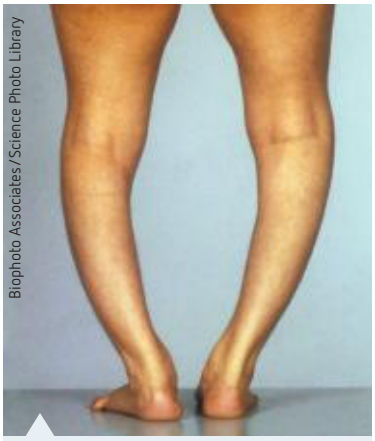


FIGURE 15.15 Bowed legs are a typical symptom of rickets, a disease caused by a prolonged lack of vitamin D in the diet of young children.

Numerous B group vitamins are necessary for proper functioning of the body. One of these is *vitamin B1*, which is responsible for growth, carbohydrate and amino acid metabolism and functioning of the heart, nerves and muscles. Prolonged deficiency of vitamin B1 in a child's diet causes a disease called **beriberi**, which results in retarded growth, weakened heart muscle, loss of appetite, confusion, inflammation of the nerves, poor coordination, tingling and paralysis.

Scurvy is a disease caused by insufficient vitamin C (ascorbic acid) in the diet. This disease causes poor wound healing, joint pains, bleeding gums, bones that do not grow or heal and spontaneous haemorrhaging. If left untreated for an extended period of time, death will result.

Lack of minerals

Iron is a mineral required by the body as an essential component in the molecule haemoglobin, which is responsible for carrying oxygen around the body. A deficiency of iron in the diet is the most common nutritional deficiency and will cause iron deficiency **anaemia**. Symptoms of iron deficiency anaemia include pale skin, weakness, unusual tiredness, apathy, low resistance to cold temperatures and difficulty breathing when exerting the body.

Iodine deficiency is another mineral nutritional disease that has re-emerged in the past few years. This could be partly attributable to the fact that dairy products no longer contain iodine and as a nation

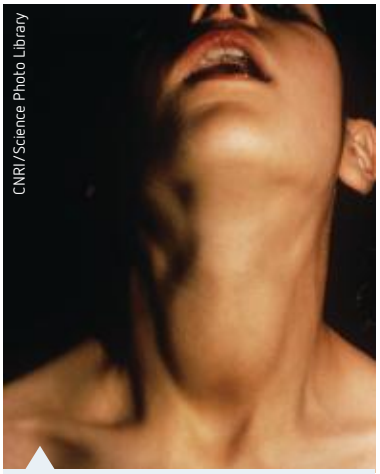


FIGURE 15.16 The characteristic swelling of the neck, called a goitre, caused by iodine deficiency

we are consuming less iodised salt. Because iodine is required for the production of thyroxin, a deficiency of iodine causes reduced metabolic rate, low body temperature and lethargy. Prolonged iodine deficiency also causes enlargement of the thyroid gland, resulting in a visible lump, known as a **goitre**, in the throat region (Fig. 15.16). Severe iodine deficiency during pregnancy causes abnormal foetal development, resulting in severely stunted mental and physical growth.

Iodine is found in fish (including tinned fish), shellfish and eggs. Also, all Australian bakers are required to use iodised salt when baking bread.

Eating disorders

Diseases that have a complex psychological basis and lead to an inappropriate diet and severe undernutrition include such conditions as **anorexia nervosa** and **bulimia nervosa**. These disorders are becoming more common in both females and males. Sufferers experience an intense fear of putting on weight. The causes vary from person to person and could be influenced by genetic predisposition and a combination of environmental, social and cultural factors. Sufferers may see themselves as being overweight when in fact they are underweight.

Anorexia nervosa is characterised by psychological disorders, excessive weight loss and a distorted body image. Sufferers often can think of nothing but their body weight and the ways in which they can keep it to a minimum. They often develop an irrational fear of both food and eating, and besides following an inadequate diet with reduced energy intake, will find other ways to lose what they perceive as excess weight. These abnormal behaviours can include excessive strenuous exercising, purging (induced vomiting), and using appetite suppressants, diuretics and laxatives.

Worksheet
Diseases caused by nutritional deficiency

The effects are loss of weight, tiredness, anaemia, impaired digestive function, bruising and low resistance to disease as well as infertility. An inadequate diet with a reduced energy and mineral intake, if left untreated, could lead to organ failure and death.

Bulimia nervosa is caused by psychological factors similar to those of anorexia nervosa, where the sufferer is fixated on body image and demonstrates abnormal eating behaviours. Bulimia is characterised by periods of binge eating followed by some method of purging, such as self-induced vomiting. The acid in the vomit causes erosion of tooth enamel, leading to erosion, sensitivity, discolouration and possible loss of teeth. The effects on the body are similar to those of anorexia nervosa, but the person does not usually become as extremely thin and emaciated. The most common cause of death in people with severe anorexia nervosa or bulimia is cardiac arrest, often due to an electrolyte imbalance.

Overnutrition

The most common type of overnutrition is obesity. Obesity is a problem in many developed countries. The cause is consuming more kilojoules than the energy expended, resulting in an accumulation of fat in the body. The overconsumption of food could be due to a number of factors, including psychological, cultural and economic factors. The body mass index (BMI) is a measure of a person's total body fat and can be used to determine whether the person is within the normal weight range, overweight or obese. An adult with a BMI over 30 can be considered obese (Fig. 15.17). BMI does not distinguish between excess fat and muscle, so elite athletes with large muscle mass can have a very high BMI.

Obesity can have a number of adverse health effects on an individual, including increased blood pressure, atherosclerosis, gallbladder disease, stroke, type 2 diabetes, problems with weight-bearing joints, increased risk of developing certain cancers such as colorectal cancers, and reduced life expectancy.

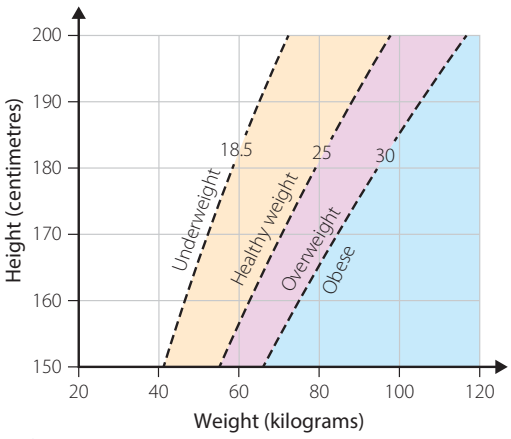


FIGURE 15.17 Body mass index (BMI) is a measure of the total body fat of an individual.

Diets that are high in certain nutrients and low in others can increase the risk of developing certain conditions. For example, a diet that is high in trans fats, saturated fats and salt, and low in fruit, vegetables and fish increases the risk of developing cardiovascular disease.

KEY CONCEPTS

- Proper nutrition contributes to better infant, child and maternal health, a stronger immune system and a reduced risk of developing a non-infectious disease.
- Nutritional diseases are caused by a diet lacking the proper balance and amount of nutrients.
- An imbalance in the diet leads to a condition known as malnutrition.
- The two broad categories of malnutrition are:
  - undernutrition – insufficient intake of the correct type of food, quantity of food or both
  - overnutrition – excessive intake of food.
- Undernutrition includes lack of protein and energy-rich foods, vitamin and mineral deficiency and eating disorders.
- Obesity is the most common form of overnutrition and can have many adverse health effects, including increased blood pressure, atherosclerosis, gallbladder disease, stroke, type 2 diabetes, problems with weight-bearing joints, increased risk of developing certain cancers such as colorectal cancers, and reduced life expectancy.

CHECK YOUR UNDERSTANDING 15.1c

1 How does a balanced diet contribute to the wellbeing of an individual?
2 **a** Define 'malnutrition'.
  b What are the two main categories of malnutrition?
3 Describe the causes and effects of both kwashiorkor and marasmus.
4 **a** Identify two vitamin deficiency diseases.
  b Describe the cause and the effects on the body of each disease you identified in **a**.
5 Construct a table to summarise the sources of the vitamins and minerals required by the body to ensure health and wellbeing.
6 Outline the cause and effects of anorexia nervosa on the body.
7 Outline the effects of obesity on the body.

Cancer

Cancer is a disease of the cells of the body. It occurs when abnormal cells divide in an uncontrolled way. These cells can invade body tissue near them, spread to other tissues and disrupt the normal functioning of the body.

Cell division is controlled by a particular set of genes contained in the DNA of the cell. These genes code for the proteins that control the correct functioning of the process of cell division. There are three types: DNA repair genes, proto-oncogenes and tumour suppressor genes.

- **DNA repair genes** code for proteins that are responsible for stopping the cell cycle while other proteins remove damaged regions of DNA and replace them with the correct sequence. If these genes mutate, they will no longer function correctly, the DNA will not be repaired, and the damaged DNA will be replicated. As a result, the correct proteins/enzymes necessary for normal cell functioning, growth and repair will not be produced and disease will occur.
- **Proto-oncogenes** code for proteins that stimulate cell growth and mitosis. Mutations of proto-oncogenes lead to the expression of **oncogenes** that would normally be 'silent'. This causes uncontrolled production of cells and prevents cell death.
- **Tumour suppressor genes** code for proteins that slow down or stop cell growth and mitosis. These genes also code for proteins that induce cell death if there is an uncontrolled increase in cell numbers. Mutations of tumour suppressor genes halt the production of proteins that control cell division and cell death.

The cell cycle is dealt with in Chapter 3, page 83.

In a healthy cell, DNA repair genes, proto-oncogenes and tumour suppressor genes tightly regulate cell growth and mitosis. If these genes are damaged or mutated, this regulation is disrupted.

Weblink
Cell cycle
Watch the video to further understand the genes responsible for regulating the cell cycle.

This disruption of the normally regulated cell cycle leads to uncontrolled cell replication, which:

- does not allow cells to differentiate, so they cannot perform the specialised functions necessary for normal body functioning
- causes the formation of tumours. If the tumour is malignant (invasive or spreading), cancer results.

One particular type of breast cancer is particularly aggressive, difficult to control and harder to treat than most forms of this disease. BRCA1, a tumour suppressor gene found on chromosome 17 (Fig. 15.18), is believed to be responsible for coding for proteins involved in the repair of the PTEN gene. Mutations of the BRCA1 gene put women at a much higher risk of developing this form of breast cancer.

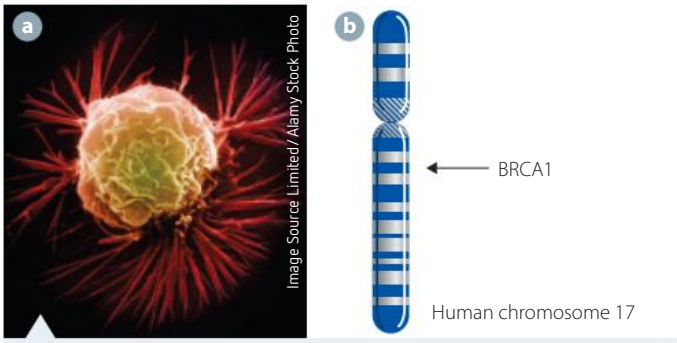


FIGURE 15.18 Mutation of the BRCA1 gene increases the risk of breast cancer: **a** EM image of a breast cancer cell; **b** position of the BRCA1 gene on chromosome 17

The PTEN gene is a tumour suppressor gene that limits the amount of cell division and encourages cell death. This regulates the cell cycle and prevents the excessive proliferation of cells that would lead to tumours and cancer.

In a normal cell, if there was damage to the PTEN gene, proteins that have been coded for by the BRCA1 gene would repair it. This repaired PTEN gene would then be expressed properly and cell division would be controlled.

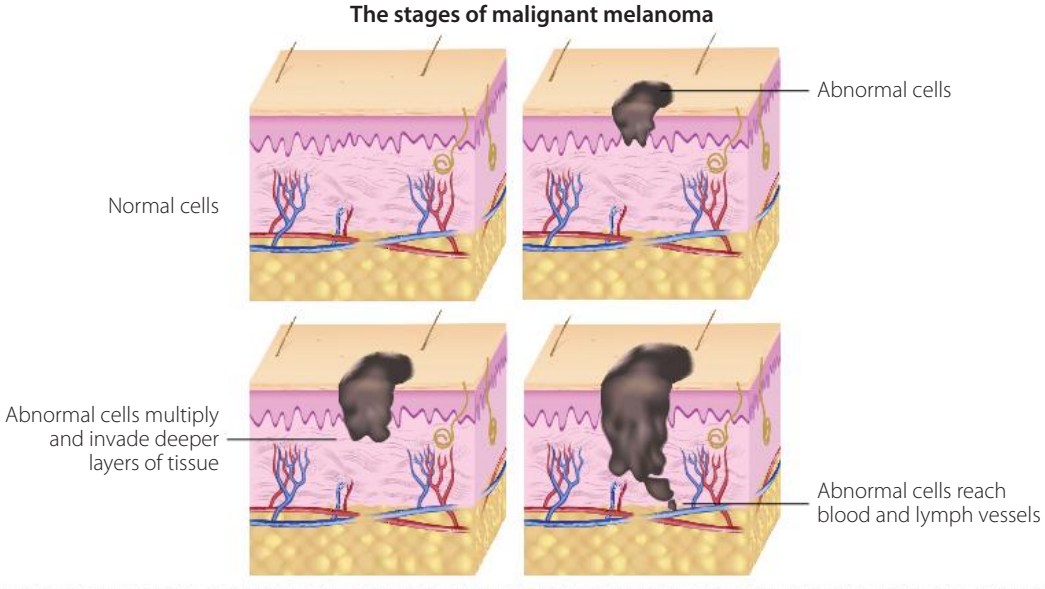
Mutation of the BRCA1 gene results in non-production of the proteins necessary for repairing damage to the PTEN gene, and so the PTEN gene remains damaged. This results in a lack of control of the cell cycle and runaway cell division, leading to the formation of tumours.

Types of tumours

Benign tumours are not classed as cancer. Their cells remain within the boundary of the tumour and do not spread to other body tissues. Some benign tumours can be precancerous and if not treated may develop into cancer. For example, squamous cell carcinoma, a type of skin cancer, can begin as a lesion (damaged area) and will spread if left untreated.

Malignant tumours contain abnormal cancerous cells that are not confined by the boundary of the initial tumour. Their cells replicate more quickly and in a more disorganised manner than normal cells, redirect the nutrients to themselves and away from adjacent normal tissue, and can invade surrounding tissue (Fig. 15.19). The area in the body where the initial malignant tumour develops is called the **primary tumour**.

FIGURE 15.19 The process of forming a malignant tumour (cancer) – in this case, a skin cancer.



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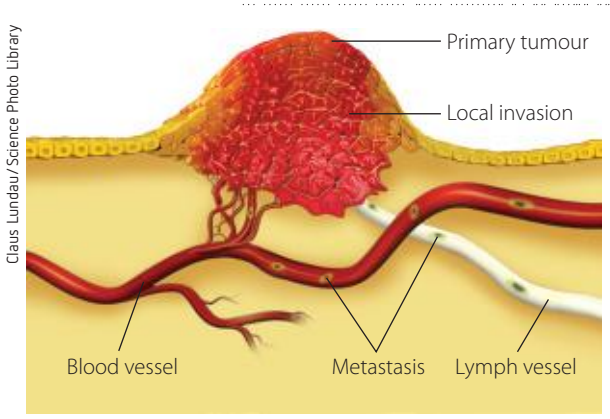
Some of these abnormal cells can break away from the tumour, enter the blood or lymphatic system and travel to a new part of the body, to develop into more tumours, known as **secondary tumours**. This process of spreading to other parts of the body is called **metastasis** (Fig. 15.20).



Claus Lundau/Science Photo Library

FIGURE 15.20 The process of metastasis – how cancer spreads. Angiogenesis, the development of new blood vessels, provides the abnormal cancerous cells with nutrients.

Types of cancer

The type of cancer is usually named after the organ or tissue that the primary tumour forms in. For example, breast cancer develops first in the breast, thyroid cancer arises in the thyroid gland.

The types of cancer can be classified into several broad groups.

- **Sarcoma** forms in muscle, or connective tissue such as bone or blood vessels.
- **Carcinoma** forms in epithelial tissue, such as skin or tissue that lines or covers the internal organs.
- **Lymphoma** and **myeloma** form in the lymphatic system and plasma cells of the immune system.
- **Leukaemia** forms in the bone marrow and other blood-forming tissues.
- **Central nervous system cancers** begin in the brain or spinal cord.

Causes of cancer

There is ongoing major research into the causes of cancer, but our understanding is still limited. It has been shown that there are a number of chemical, physical and biological agents, known as carcinogens (such as tobacco, UV radiation and asbestos), that could cause mutations to the genes that control cell division.

Worksheet
The role of genes in the development of cancer

In many cases, there may be not just one risk factor but a combination of risk factors involved in the occurrence of cancer.

Other risk factors associated with the occurrence of cancer are:

- smoking – one in nine cancers and one in five deaths from cancer are associated with smoking
- excessive alcohol consumption
- lack of physical activity, being overweight or obese and following a diet that is high in saturated fats
- exposure to radiation, such as UV radiation
- contracting particular types of viruses, such as the human papilloma virus (HPV) or the hepatitis B or C virus – HPV is associated with cervical cancer and other cancers, while hepatitis B and C can be associated with liver cancer
- exposure to certain chemicals in the workplace, home or environment, such as asbestos and organic solvents
- inheriting mutated genes that predispose a person to developing certain types of cancers – for example, the BRCA1 gene increases the likelihood of developing breast cancer by 85%.

Some risk factors, such as those that are related to lifestyle and the environment, can be minimised. Others, such as those associated with heredity and biological factors, cannot. Some people with one or more risk factors do not develop cancer, while others with no known risk factors do develop cancer.

Melanoma: a form of skin cancer

Melanoma is a disease in which cells in the skin divide uncontrollably due to changes in the DNA of the genes that control cell division. These cells are called melanocytes and contain the pigment melanin, which gives the skin its colour. Melanoma can occur anywhere on the skin, but in men it is more common on the head, neck and trunk, and in women it is more common on the legs and arms.

The exact cause of melanoma is not known, but the chance of developing this cancer is increased by the following risk factors:

- exposure to UV light from the sun or a tanning bed
- fair complexion, blue or green eyes, blonde or red hair, freckles, skin that burns easily
- severe sunburn that caused blistering, in childhood
- a history of having skin cancer, including melanoma
- having many different types of moles or irregular-looking moles
- a family history of moles or melanoma
- exposure to certain environmental conditions or chemicals, such as radiation or organic solvents
- a compromised immune system
- older age – being older increases the risk of developing melanoma
- male gender – men are more likely to develop melanoma than women.

Weblink
Melanoma
Outline the cells that are affected by melanoma and how they become cancerous.

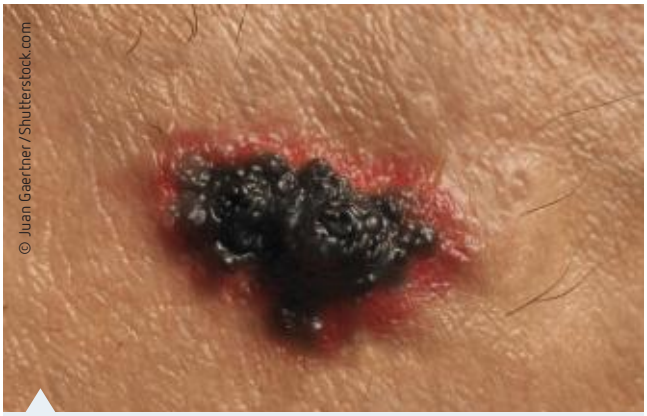


FIGURE 15.21 Melanoma is formed by the uncontrolled division of the cells in the skin that produce the brown pigment melanin.

The malignant melanoma tumour initially is confined to one area but if left untreated will increase in thickness, spreading to deeper layers within the skin. With the further passage of time, the melanoma will spread to nearby lymph nodes, lymph vessels and skin. The cancerous cells will then travel to distant lymph nodes, other areas of the skin and other organs such as the lungs, brain, bones and liver. This will interfere with the correct functioning of the body and unless successfully treated will lead to death.

INVESTIGATION 15.1

A secondary-source investigation into the causes and effects on the body of non-infectious disease

Information and communication technology capability

There are many different types of non-infectious diseases, and their causes vary. Non-infectious diseases include genetic diseases, diseases caused by environmental factors, nutritional diseases and cancer. Some of these diseases are currently the leading causes of death in Australia (Table 15.1, page 506).

AIM

To investigate the cause and effect on an individual of one disease in each of the following categories: genetic disease, environmental disease, nutritional disease, cancer

»

METHOD

1. Revise the information presented in this chapter about the different types of non-infectious disease.
2. Work collaboratively in groups to research information about another example of a disease from each of the categories listed in the aim.
3. Use a wide range of secondary sources and assess the relevance, accuracy, validity and reliability of each source.
4. Identify the category of the disease, the name of the disease, the cause of the disease and the effect it has on an individual.
5. Work collaboratively in a Google Doc, or collate results from each member of the team, and present your results in a manner of your choosing – for example, in a table like the one supplied in the Results section.
6. Acknowledge your sources using an accepted referencing style.

RESULTS

Use a table like the one below to record your results.

	CATEGORY OF NON-INFECTIOUS DISEASE			
	GENETIC	ENVIRONMENTAL	NUTRITIONAL	CANCER
Name of disease				
Cause of disease				
Effects of the disease on the individual				

DISCUSSION

1. Define 'non-infectious disease'.
2. Differentiate between the four categories of disease listed in the results table.
3. Assess the relevance, accuracy, validity and reliability of the sources used to obtain the information required.

Refer to Chapter 1, page 10 for guidance on assessing the relevance, accuracy, validity and reliability of secondary sources used.

CONCLUSION

Write summary sentences related to the aim of this investigation.

KEY CONCEPTS

- Cancer occurs when abnormal cells divide in an uncontrolled way.
- DNA repair genes, proto-oncogenes and tumour suppressor genes control cell division. In a healthy cell, these genes tightly regulate cell growth and mitosis. If these genes are damaged or mutated, this regulation is disrupted, which may lead to cancer.
- Benign tumours are not classed as cancer. Their cells remain within the boundary of the tumour and do not spread to other body tissues.
- Malignant tumours contain abnormal cancerous cells that are not confined by a boundary.
- The type of cancer is usually named after the organ or tissue that the primary tumour forms in.
- The types of cancer are:
  - sarcoma – forms in connective tissue or muscle (e.g. bone, blood vessels)
  - carcinoma – forms in epithelial tissue (e.g. skin, tissue lining internal organs)
  - lymphoma and myeloma – forms in the lymphatic system and immune system
  - leukaemia – forms in bone marrow and other blood-forming tissues
  - central nervous system cancers – form in the brain or spinal cord.
- Cancer is often caused by exposure to one or more risk factors.

CHECK YOUR UNDERSTANDING 15.1d

1 **a** Identify the genes involved in the regulation of the cell cycle.
   b Describe the function of each of these types of genes.
   c Outline what happens when mutations occur in:
      i proto-oncogenes
      ii tumour suppressor genes.
2 Distinguish between benign and malignant tumours, and identify which of these is called cancer.
3 Define each of the following terms:
   a primary tumour
   b secondary tumour
   c metastasis.
4 What are the main categories of cancer?
5 **a** What is a risk factor?
   b Identify common risk factors associated with developing cancer.
6 Outline the cause of melanoma and its effects on the body.

15.2 Incidence, prevalence and mortality rates

Non-infectious disease is the cause of 70% of deaths globally, with 'lifestyle' diseases responsible for 63% of these deaths. Data relating to the incidence, mortality and prevalence of many types of non-infectious disease can be collected and analysed to identify patterns in populations. The incidence, prevalence and mortality rates of diseases can be determined for whole populations and subsets of populations. The **incidence** of a disease refers to the number of new cases of that disease reported in a specific time period (usually a year). The **prevalence** of a disease is the number of people in a particular population that have been diagnosed with that disease and are still alive at the end of a given time period.

It is important to note that the prevalence of a disease is in effect how many people in the population have that disease, and the incidence is how many *new* cases of the disease are diagnosed within a given time period. For example, if a large number of new cases of a disease are diagnosed in one year, then the incidence and prevalence will both be high that year. If a method were found to prevent that disease from occurring, then the incidence the next year would be low, as there would be very few new cases, but the prevalence would still be high as there are still people with that disease from the previous year.

The **mortality rate** of a disease is the number of deaths due to a particular disease in a specific time period (usually one year). The incidence and mortality rates are usually expressed as a number per 100 000 population. The **age-standardised rate** is a measure of what the rate would be if the population had a standard age structure. It is a weighted mean (similar to an average, but the data points have different weights) of the age-specific rates and is used so that comparisons between populations from different countries or regions can be made.

This data is useful in many ways, including being used to determine:

- the trends associated with particular diseases
- whether certain groups are more susceptible to particular diseases
- whether methods in use for preventing and treating the disease are successful
- the pathway for research and areas to concentrate on in terms of public health.

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INVESTIGATION 15.2

A secondary-source investigation to analyse data on the incidence, mortality and prevalence of melanoma

Critical and creative thinking

Numeracy

Difference and diversity

Melanoma is a type of cancer that can be caused by environmental exposure to UV light (page 522). The following information about melanoma is data that has been collected on the incidence, prevalence and mortality rates for the Australia population as a whole and for different population groups in Australia. There is also data that relates to the incidence, mortality and prevalence of melanoma in the world and in different populations in the world. This data is in the form of raw data, tables and graphs.

AIM

To interact with and analyse data relating to the incidence, mortality and prevalence of melanoma

METHOD

PART A: INCIDENCE AND MORTALITY RATES OF MELANOMA IN AUSTRALIA

Read the following information and refer to Tables 15.3 and 15.4 and Figure 15.22, then answer the questions that follow.

Melanoma was the 4th most commonly diagnosed cancer in Australia in 2013. It is estimated that it will remain the 4th most commonly diagnosed cancer in 2017. The incidence rate of melanoma skin cancer generally increases with age.

In 2013, there were 12 744 new cases of melanoma diagnosed in Australia (7513 males and 5232 females). In 2017, it was estimated that 13 941 new cases of melanoma would be diagnosed in Australia (8392 males and 5549 females).

In 2013, the age-standardised incidence rate was 50 cases per 100 000 people (62 for males and 40 for females). In 2017, it was estimated that the age-standardised incidence rate would be 50 cases per 100 000 people (62 for males and 39 for females).

The number of new cases of melanoma diagnosed increased from 3527 (1733 males and 1794 females) in 1982 to 12 744 in 2013. Over the same period, the age-standardised incidence rate increased from 27 cases per 100 000 people (28 for males and 26 for females) in 1982 to 50 cases per 100 000 people in 2013.

The above information about the total number of new cases of melanoma, along with the values for males and females, and the age-standardised incidence rates, has been summarised in Table 15.3.

TABLE 15.3 Incidence of melanoma in Australia

YEAR	NUMBER OF NEW CASES			AGE-STANDARDISED INCIDENCE RATE (PER 100 000)		
	TOTAL	MALE	FEMALE	TOTAL	MALE	FEMALE
1982	3 527	1 794	1 733	27	28	26
2013	12 744	7 513	5 232	50	62	40
2017*	13 941	8 392	5 549	50	62	39

* estimated

Source: Australian Cancer Incidence and Mortality 2016. Australian Institute of Health and Welfare. Creative Commons BY 3.0 (CC-BY 3.0) licence.

At the time of writing, only the estimated incidence and mortality rates for 2017 were available. These are based on the patterns demonstrated in previous years and it is assumed that these patterns will continue. Actual incidence and mortality rates may vary for a number of reasons, which could include better methods of detection and diagnosis. Improved treatment options could also reduce the mortality rate.

TABLE 15.4 Incidence of melanoma by age group

	0–39	40–49	50–59	60–69	70–79	80+
Year	Number of new cases					
1982	1000	588	675	673	429	154
1992	1367	1100	1055	1435	1116	493
2002	1396	1526	1987	1865	1939	1113
2012	1193	1411	2239	2879	2435	1878
2016*	1221	1491	2443	3247	2714	2168
	Age-specific rate (per 100 000)					
1982	10.1	36.4	44.6	57.1	62.2	57.9
1992	12.7	46.0	65.6	102.2	121.0	122.5
2002	12.7	53.1	84.0	121.7	168.0	180.2
2012	9.8	44.6	77.5	128.1	183.4	215.8
2016*	9.4	45.4	80.2	129.7	174.7	229.1

* estimated

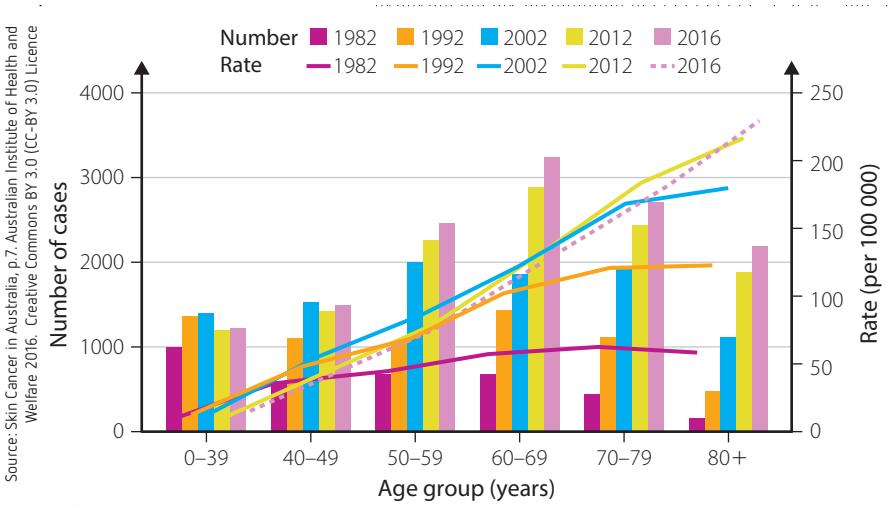

FIGURE 15.22 Number of new cases and age-specific incidence rates for melanoma, by age group, 1982, 1992, 2002, 2012 (actual data) and 2016 (estimated data)

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DISCUSSION

1. Consider the presentation of information about the incidence and incidence rate of melanoma. Is it easier to interpret written information or information presented in tabular form?
2. Using which form of information – written, tabular or graphical – is it easier to draw conclusions about the trends shown in the data?
3. What is the difference between the terms 'incidence' and 'incidence rate'?
4. Refer to Table 15.3.
   - a. What was the total number of new cases of melanoma estimated to be diagnosed in 2017?
   - b. What percentage of those cases are:
     - i. male
     - ii. female?
5. Make a statement that identifies the pattern for incidence and incidence rate for melanoma in males compared to females in all years presented.
6. Compare the total incidence rate for melanoma for each year.
7. Referring to Table 15.3, describe the pattern shown for the overall incidence rate of melanoma from 1982 to 2017.
8. Refer to Table 15.3, Table 15.4 and Figure 15.22.
   - a. For 2016:
     - i. In which age group was the highest number of new cases of melanoma diagnosed?
     - ii. Which age group had the highest age-specific incidence rate?
     - iii. Which age group had the lowest age-specific incidence rate, and what was the % difference in the rate between the two age groups?
   - b. For 1982–2016:
     - i. Describe the trend in the number of new cases of melanoma diagnosed in the age groups 50–59 and above.
     - ii. Describe the trend in the number of new cases of melanoma diagnosed in the age groups 0–39 and 40–49.

PART B: MORTALITY RATES OF MELANOMA IN AUSTRALIA

Read the following information and refer to Table 15.5, Figures 15.23 and 15.24, then answer the questions that follow.

In 2014, melanoma was the 10th leading cause of cancer deaths in Australia. It is estimated that it will become the 8th most common cause of death from cancer in 2017. The mortality rate of melanoma will generally increase with age.

In 2017, it is estimated that the risk of an individual dying from melanoma skin cancer by their 85th birthday will be 1 in 119 (1 in 77 males and 1 in 227 females).

The number of deaths (total, males and females) from melanoma along with the age-standardised mortality rates for each in 1968, 2014 and 2017 have been summarised in Table 15.5.

TABLE 15.5 Mortality rates for melanoma in Australia

YEAR	NUMBER OF DEATHS			AGE-STANDARDISED MORTALITY RATE (PER 100,000 PERSONS)		
	TOTAL	MALE	FEMALE	TOTAL	MALE	FEMALE
1968	315	178	137	3.3	3.9	2.8
2014	1467	988	479	5.5	8.1	3.3
2017*	1839	1280	559	6.3	9.5	3.6

* estimated

Based on: Skin Cancer in Australia. Australian Institute of Health and Welfare 2016. Creative Commons BY 3.0 (CC-BY 3.0) Licence

The graph in Figure 15.23 shows the patterns associated with both the age-standardised incidence rate and the mortality rate for melanoma in total and for males and females individually.

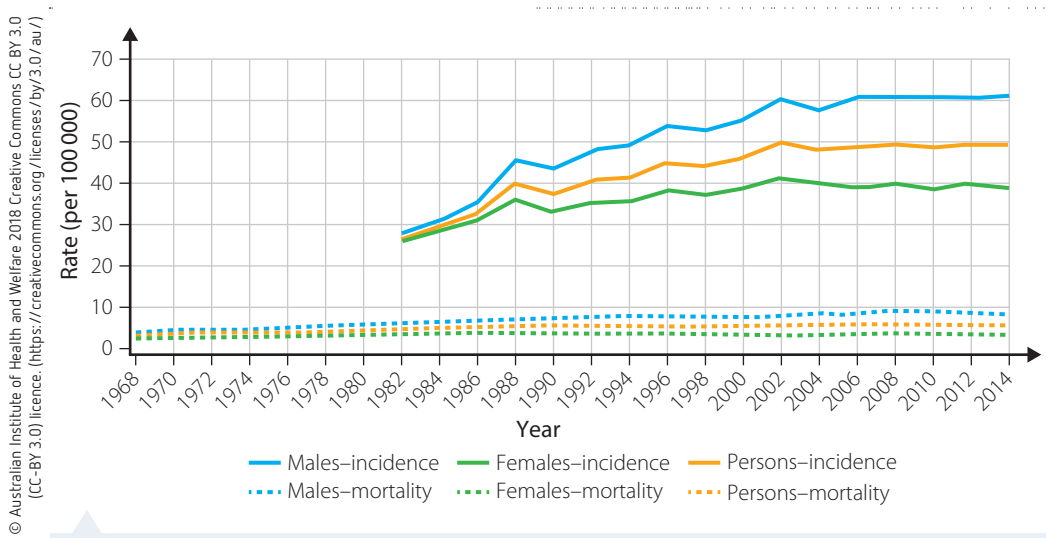

FIGURE 15.23 Age-standardised rates per 100 000 for the incidence (1982–2014) and mortality (1968–2014) of melanoma

The age-specific incidence and mortality rate per 100 000 by age group, for melanoma, is shown in Figure 15.24. The rates for all persons, males and females have been included.

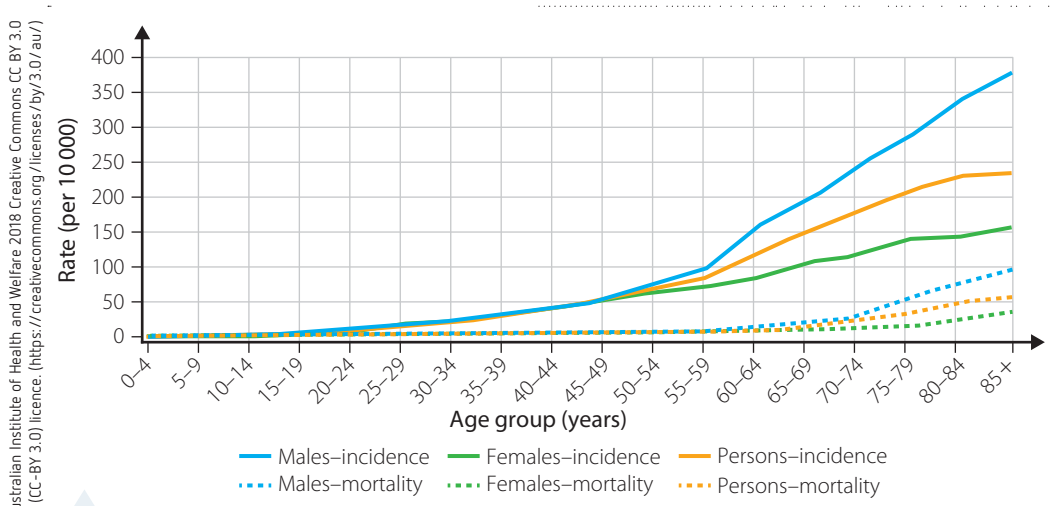

FIGURE 15.24 Estimated age-specific incidence and mortality rates per 100 000 for melanoma, 2017

DISCUSSION

Use information from Table 15.5, Figures 15.23 and 15.24 to answer the following questions.

1. Define 'mortality rate'.
2. Outline the trend in mortality rates in the years 1968–2017 and the difference between males and females.

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» **3 a** Copy and complete the following table using information from Table 15.5.

TIME PERIOD	INCREASE IN AGE-STANDARDISED MORTALITY RATE (%)		
	TOTAL	MALES	FEMALES
1968–2014			
2014–2017			

b What does this information indicate about the increase in mortality rates for melanoma in males and females?

4 Referring to Figure 15.24, outline the relationship between mortality rate and age, overall and for males and females separately.

Part C: Population groups within Australia

Aboriginal and Torres Strait Islander people

The data in Table 15.6 lists the incidence and age-standardised incidence rates for melanoma, for both Indigenous and non-Indigenous groups in Australia for the period 2005–2009.

TABLE 15.6 Number of new cases diagnosed and age-standardised incidence rates for melanoma, by Indigenous status, 2005–2009

INDIGENOUS STATUS	NUMBER OF NEW CASES DIAGNOSED	AGE-STANDARDISED RATE (PER 100 000)
Aboriginal and Torres Strait Islander	84	9.3
Non-Indigenous	22 328	33.0
Not stated*	14 906	N/A
Total	37 318	54.3

* A limitation of this data is that in 40% of cases the Indigenous status was not stated.
N/A: not applicable

Source: Skin Cancer in Australia, p.27. Australian Institute of Health and Welfare 2016. Creative Commons BY 3.0 (CC-BY 3.0) Licence

States and territories of Australia

Australia is a very large continent and has a number of climatic zones, ranging from tropical and subtropical areas in the north to much colder areas in the south. Table 15.7 summarises the incidence and age-standardised incidence rates for melanoma in the different states and territories of Australia in the period 2005–2009.

TABLE 15.7 Number of new cases diagnosed and age-standardised incidence rates for melanoma, by state or territory, 2005–2009

STATE OR TERRITORY	NUMBER OF NEW CASES	AGE-STANDARDISED INCIDENCE RATE (PER 100 000)
New South Wales	17 986	49.0
Victoria	11 259	41.2
Queensland	13 882	66.7
Western Australia	5 201	49.1
South Australia	3 381	37.9
Tasmania	1 346	48.9
Australian Capital Territory	672	42.3
Northern Territory	249	32.3
Total	53 976	49.3

Source: Skin Cancer in Australia, p.28. Australian Institute of Health and Welfare 2016. Creative Commons BY 3.0 (CC-BY 3.0) Licence

DISCUSSION

1 What percentage of the total number of new cases of melanoma in 2005–2009 was diagnosed in:
   a Aboriginal and Torres Strait Islander Australians?
   b non-Indigenous Australians?

2 Compare the age-standardised incidence rate for melanoma of Aboriginal and Torres Strait Islander and non-Indigenous Australians from 2005 to 2009.

3 a What is one limitation of the data presented in Table 15.6?
   b How would this limitation affect the data collected?

4 Compare the incidence and age-standardised incidence rate for melanoma by state or territory for 2005–2009.

5 Explain why NSW has the highest number of new cases of melanoma but not the highest age-standardised incidence rate.

Part D: Incidence of melanoma in different countries of the world

Worldwide

The age-standardised incidence rates for melanoma in different countries of the world are shown in Figure 15.25.

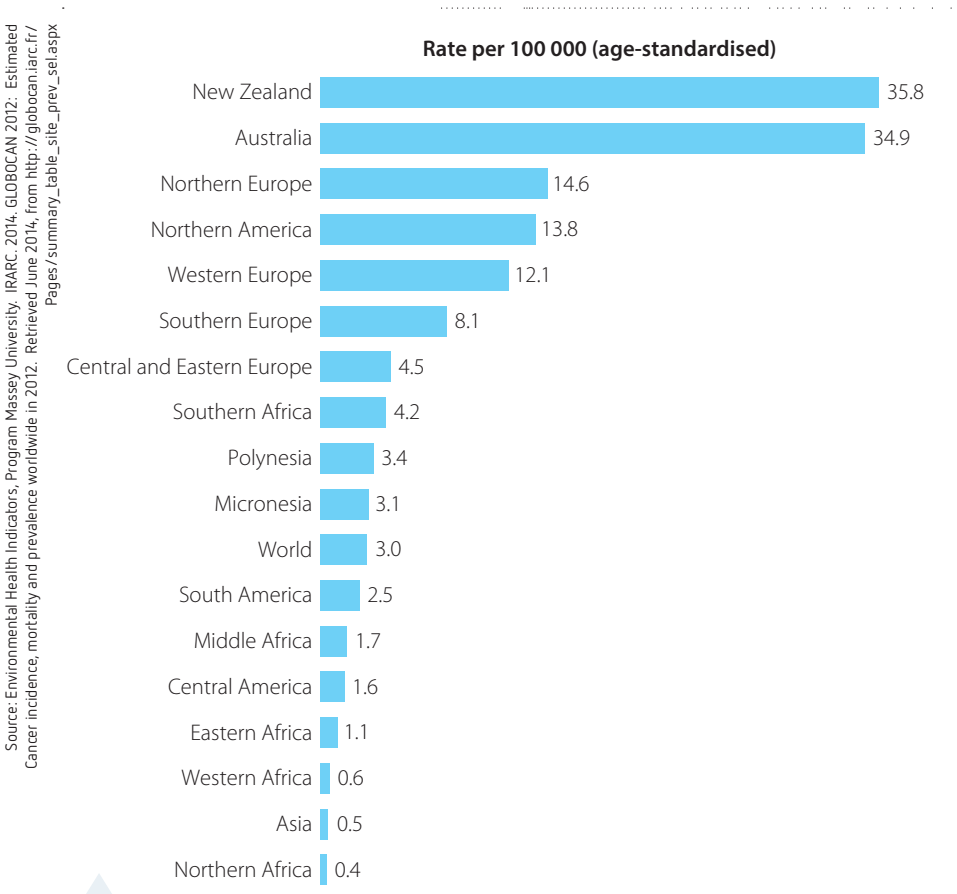


FIGURE 15.25 Melanoma incidence rate, countries and regions of the world, 2012

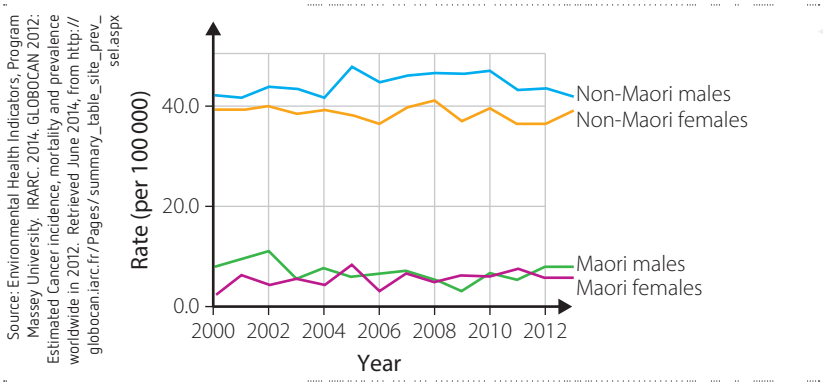


FIGURE 15.26 Age-standardised incidence rate (per 100 000) for melanoma 2000–2012, for Non-Māori and Māori males and females in New Zealand

USA

The graphs in figures 15.27 and 15.28 represent the age-standardised incidence rate of melanoma by race and ethnicity for males and females respectively from 1999 to 2014.

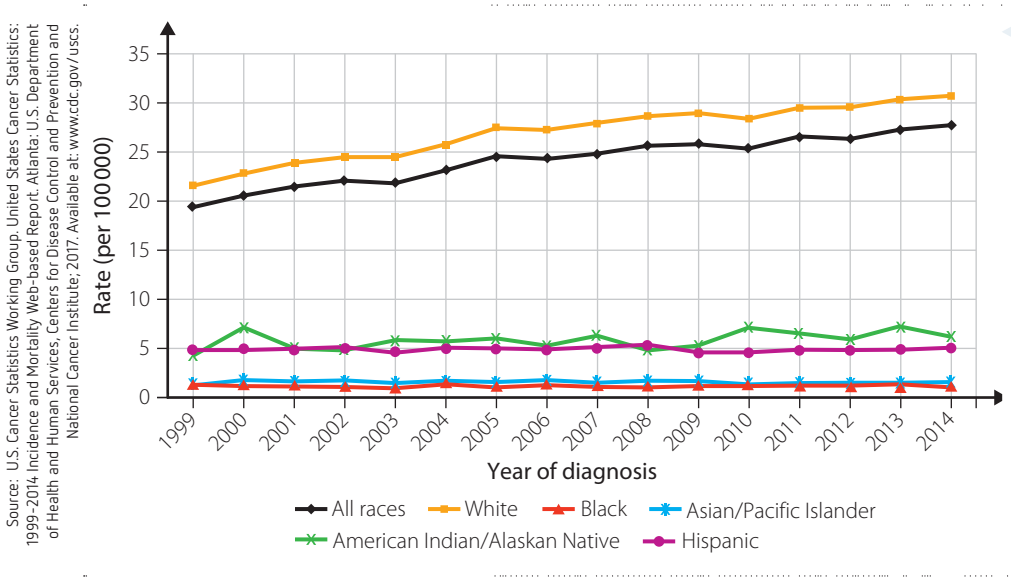


FIGURE 15.27 Age-standardised incidence rates for melanoma in males of different races and ethnicities in the USA for 1999–2014

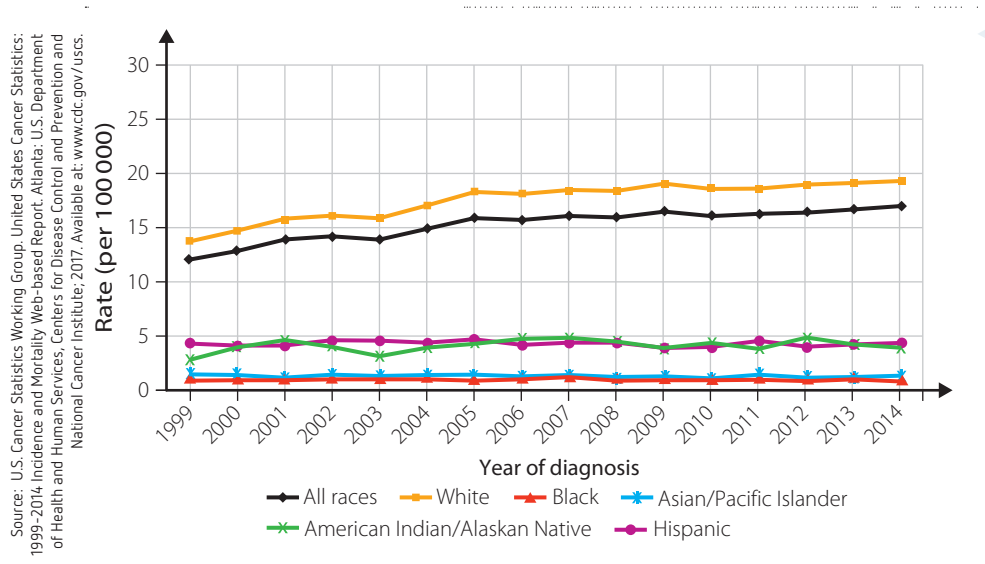


FIGURE 15.28 Age-standardised incidence rates for melanoma in females of different races and ethnicities in the USA for 1999–2014

DISCUSSION

1 Which country or region of the world has the:
   a highest age-standardised incidence rate of melanoma in the world
   b lowest age-standardised incidence rate of melanoma in the world?

2 Identify and describe the common trends and patterns shown by the data in figures 15.26,15.27 and 15.28.

Part E: Prevalence of melanoma

The prevalence of melanoma in the Australian population at the end of 2012 is shown in Table 15.8. The number of people still alive at the end of 2012 who had been diagnosed with melanoma in 2012 was 11 960. The number of people still alive at the end of 2012 who had been diagnosed with melanoma between 2008 and 2012 was 51 697, and the number of people still alive at the end of 2012 who had been diagnosed between 1982 and 2012 was 169 292.

TABLE 15.8 Prevalence of melanoma in the Australian population

YEAR DIAGNOSED	NO. PEOPLE LIVING AT THE END OF 2012 WHO HAD PREVIOUSLY BEEN DIAGNOSED WITH MELANOMA
2012	11 960
2008–2012	51 697
1982–2012	169 292

Source of data: Australian Government, Cancer Australia, 2018, 'Melanoma of the skin statistics', https://melanoma.canceraustralia.gov.au/statistics

The prevalence of a specific disease can be used to calculate the survival rate associated with that disease.

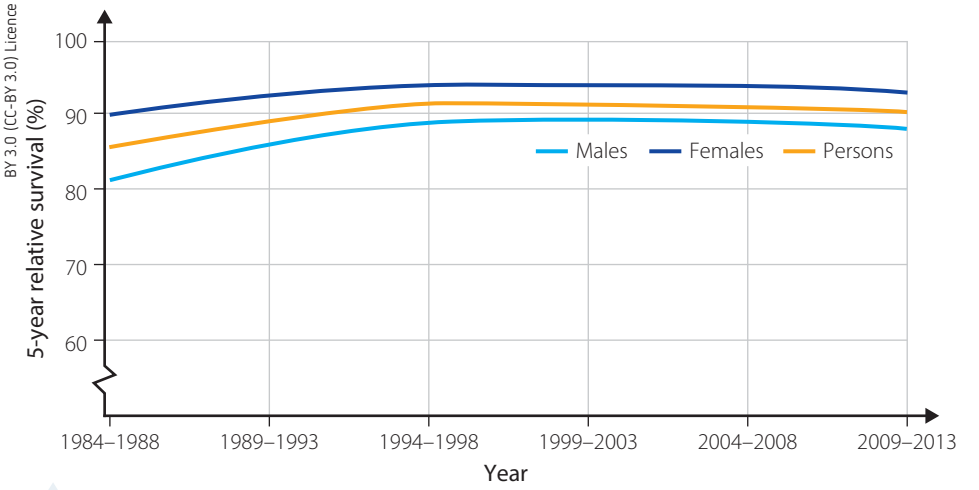


FIGURE 15.29 Five-year relative survival from melanoma 1984–1988 to 2009–2013

It was estimated that, in 2017, the risk of an individual being diagnosed with melanoma skin cancer by their 85th birthday would be 1 in 17 (1 in 13 males and 1 in 23 females).

The incidence and prevalence of melanoma for males and females in different regions of the world in 2012 are shown in Fig 15.30.

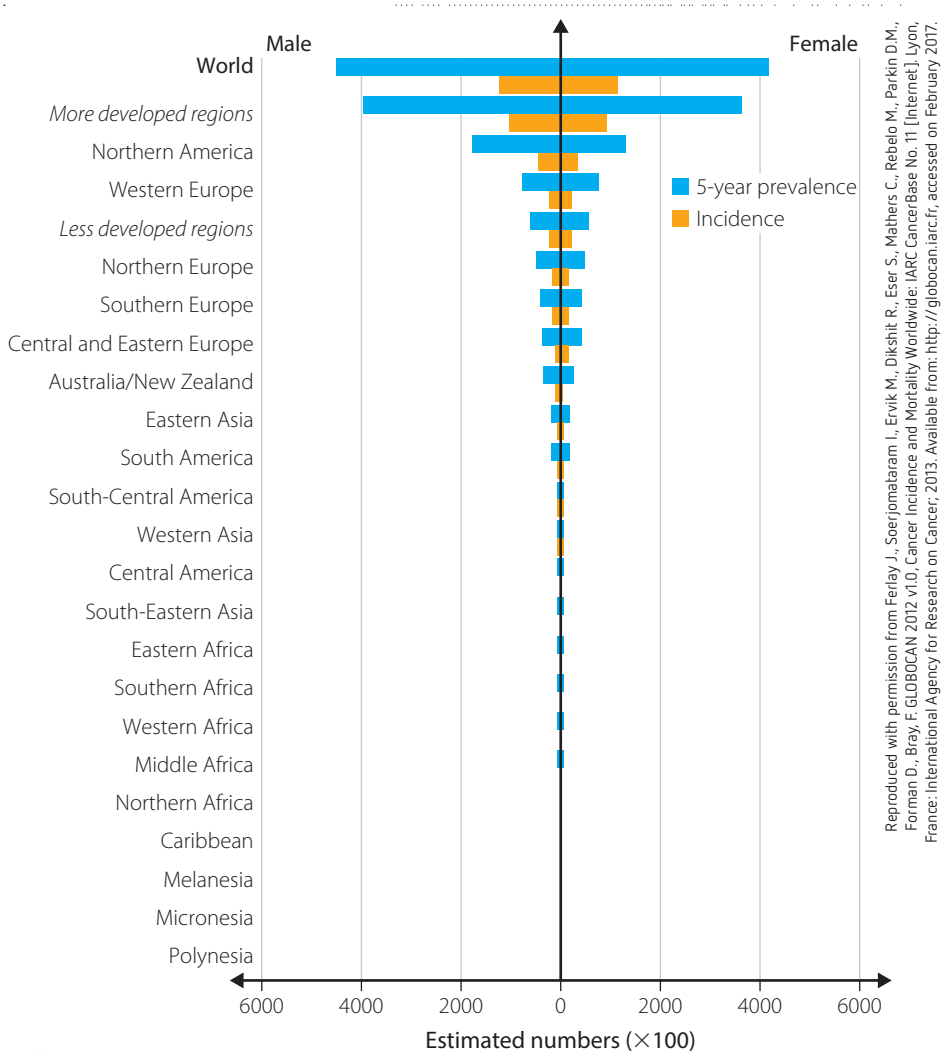


FIGURE 15.30 Five-year prevalence and incidence data for the world and regions of the world for melanoma in 2012

DISCUSSION

1. What was the prevalence of melanoma in 2012 for those diagnosed in the time period:
   - **a** 1982–2012
   - **b** 2008–2012
   - **c** 2012?
2. Compare the 5-year survival rate for the total population and for males and females in 2009–2013.
3. Compare trends in incidence and prevalence of melanoma between the more developed and less developed regions of the world.
4. Form hypotheses to explain why, in Australia:
   - **a** there has been a relatively constant incidence rate of melanoma since 2013
   - **b** the incidence and incidence rate of melanoma in males are higher than in females.
5. Suggest reasons why the mortality rate from melanoma is so much higher in males than in females.

6 Explain how the data in Table 15.6 supports the statement: 'Aboriginal and Torres Strait Islander Australians have a lower risk of developing melanoma because of the high level of skin pigment in their cells'.

7 Relate the data in Table 15.7 to the statement :'Tropical and subtropical areas in the north of Australia have a much greater exposure to higher levels of UV radiation from the sun than the much colder areas in the south of the country'. Account for any anomalies noted in the data, particularly in regard to the Northern Territory.

8 Provide hypotheses as to why Australia and New Zealand have a higher incidence rate of melanoma than northern Europe and northern America, and why Central America has a much lower incidence rate than northern America even though it has a much greater exposure to UV light.

9 Suggest reasons why females diagnosed with melanoma have a higher 5-year relative survival chance than males.

10 Construct evidence-based arguments that could be presented to adolescents to warn them of the dangers of melanoma and the risk factors involved.

CONCLUSION

Make a series of statements related to the patterns of incidence, mortality rate and prevalence of melanoma. These statements should include factors such as age, gender, race and geography.

KEY CONCEPTS

- Non-infectious disease is the cause of 70% of deaths globally, with 'lifestyle' diseases being responsible for 63% of these deaths.
- The incidence, prevalence and mortality rates of diseases can be used in numerous ways to improve the health and wellbeing of populations.
- The incidence of a disease is the number of new cases of that disease reported in a specific time period (usually a year).
- The prevalence of a disease is the number of people in a particular population that have been diagnosed with that disease and are still alive at the end of a given time period.
- The mortality rate of a disease is the number of deaths due to that disease in a specific time period (usually one year).
- The incidence and mortality rates of melanoma increase with age.
- The incidence rate of melanoma in Australia has steadied, the mortality rate is increasing, and the rate of both is higher for males than females.
- Predominantly dark-skinned populations throughout the world have a lower incidence of melanoma than fair-skinned populations.
- Fair-skinned populations in higher latitudes, such as Australia and New Zealand, have the highest rates of melanoma in the world.
- The prevalence of melanoma is increasing in Australia and is greater in the more developed regions of the world.

CHECK YOUR UNDERSTANDING 15.2

1 a Define the following terms:
   i incidence
   ii prevalence
   iii mortality rate.
   b Distinguish between incidence and prevalence, using an example.

2 Outline ways in which data about diseases can be used to benefit the health and wellbeing of a population.

3 Outline the difference between the following terms: incidence, age-standardised incidence, age-specific incidence.

INVESTIGATION 15.3

A secondary-source investigation into the incidence, prevalence and mortality rates of nutritional diseases

There are many types of nutritional diseases, including type 2 diabetes and obesity. The incidence and mortality rate of each of these two diseases has increased in recent years and both diseases are becoming more prevalent both globally and in Australia.

Information and communication technology capability

Numeracy

Difference and diversity

AIM

To collect and represent data that shows the incidence, prevalence and mortality rates of a nutritional disease

METHOD

1. Choose a nutritional disease that has been discussed in this chapter, or another that interests you.
2. Research numerous sources to locate data about your chosen disease.
3. Collect data about the incidence, mortality rate and prevalence of the disease in Australia and other regions of the world.
4. The data should include values for males, females and any other characteristic pertaining to your chosen disease.
5. Calculate percentage differences between males and females in terms of the incidence and mortality rates, and present the information in an appropriate table.
6. Reference your sources of data and assess the relevance, reliability, validity and accuracy of these sources.
7. Include any errors in the data as well as any limitations associated with the data you have collected.

Refer to the previous section, page 525, for guidance on the type of data to collect for this investigation.

RESULTS

1. Using headings, present your data in an appropriate form, such as written, tables or graphs.
2. Assess the relevance, reliability, validity and accuracy of the sources you used to access the data you collected.

Refer to Chapter 1 for information about referencing (page 27) and assessing the relevance, reliability, validity and accuracy (page 10) of sources.

DISCUSSION

1. Define the following terms:
   a. incidence
   b. mortality
   c. prevalence.
2. Outline the difference between the following: incidence, age-standardised incidence, age-specific incidence.
3. Suggest reasons why it is important to the health and wellbeing of a population that data such as this be collected and analysed.

CONCLUSION

Provide summary sentences related to the aim of this investigation.

INVESTIGATION 15.4

A secondary-source investigation to determine whether non-infectious diseases cause more deaths than infectious diseases

INTRODUCTION

Numeracy

Historically, infectious disease has been the leading cause of death in the world. With the advent of improved measures to prevent and treat infectious diseases, non-infectious disease is now the leading cause of death globally. However, even though non-infectious diseases caused 70% of deaths globally in 2015, if the statistics are examined on an economy-income basis, it can be seen that, in countries with a low income, infectious diseases are still the leading cause of death.

Additionally, data on the leading causes of death shows that the causes vary, depending on the age group and the gender being examined. For example, the leading cause of death for males aged 15–29 years was road injury, whereas maternal conditions were the leading cause of death for females in the same age group.

AIM

To determine whether non-infectious disease causes more deaths than infectious disease

METHOD

1 Refer to tables 15.9 and 15.10, which list the top 10 causes of death globally in 2015 and 2000 respectively. In the '% of total' column in each table, calculate the percentage figure for deaths from each cause. The total number of deaths in 2015 was 54.7 million and in 2000 was 56.4 million.

TABLE 15.9 Top ten causes of death globally, 2015

CAUSE OF DEATH	NUMBER (MILLIONS)	% OF TOTAL*
Ischaemic heart disease	8.76	
Stroke	6.24	
Lower respiratory infections	3.19	
Chronic obstructive pulmonary disease	3.17	
Trachea, bronchus, lung cancers	1.69	
Diabetes mellitus	1.59	
Alzheimer disease and other dementias	1.54	
Diarrhoeal diseases	1.39	
Tuberculosis	1.37	
Road injury	1.34	

* Total no. deaths (all causes), 2015: 54.7 million

Adapted from WHO, 'Health statistics and information systems. Estimates for 2000–2015. Global summary estimates, http://www.who.int/healthinfo/global_burden_disease/estimates/en/index1.html

TABLE 15.10 Top ten causes of death globally, 2000

CAUSE OF DEATH	NUMBER (MILLIONS)	% OF TOTAL*
Ischaemic heart disease	6.88	
Stroke	5.41	
Lower respiratory infections	3.41	
Chronic obstructive pulmonary disease	2.95	
Diarrhoeal diseases	2.18	
Tuberculosis	1.67	
HIV/AIDS	1.46	
Preterm birth complications	1.34	
Trachea, bronchus, lung cancers	1.26	
Birth asphyxia and birth trauma	1.12	

*Total no. deaths (all causes), 2000: 56.4 million

Adapted from WHO, 'Health statistics and information systems. Estimates for 2000–2015. Global summary estimates, http://www.who.int/healthinfo/global_burden_disease/estimates/en/index1.html

2 Construct graphs to show the trends recorded in tables 15.9 and 15.10 for the cause of death and the percentage deaths.

Refer to Chapter 1 page 21 to determine the most appropriate type of graph to draw.

Refer to Table 15.1 on page 506 listing the top 10 causes of death in Australia in 2015.

3 Draw a graph to represent this data, showing the cause of death and the percentage for each cause.

DISCUSSION

Use the information in tables 15.9 and 15.10, as well as the graphs you have drawn, to answer the following questions.

1 For all the causes of death listed in tables 15.1, 15.9 and 15.10, identify those that are:
   - **a** infectious
   - **b** non-infectious.

   Present your answer in table form.

2 **a** Identify any common trends between the three graphs you have drawn.
   - **b** Compare the top 10 causes of death:
     - **i** globally in 2015 with 2000
     - **ii** globally in 2015 with Australia 2015.
   - **c** Provide reasons for the similarities/differences in each comparison you made in part b.

3 Use the weblink and other sources to research whether these trends hold true for:
   - **a** low-income countries
   - **b** males and females
   - **c** different age groups.

Weblink
Top 10 causes of death
View the interactive data to compare the differences in causes of death in different regions of the world, different sexes and different age groups.

CONCLUSION

Provide summary sentences related to the aim of this investigation.

15 CHAPTER SUMMARY

Non-infectious diseases: Do non-infectious diseases cause more deaths than infectious diseases?

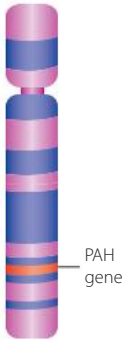
Non-infectious diseases

- Not caused by a pathogen and not contagious
- Leading cause of death globally
- Risk influenced by age, gender, economic and social conditions, culture, race, lifestyle, environment, genetics and nutrition

Genetic disease

Single-gene abnormalities

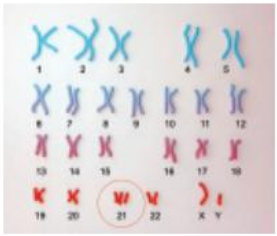
- Inherit mutated gene from parents
- Abnormal protein
- Genetic disease, e.g.
  - cystic fibrosis, haemophilia, Huntington's chorea
  - PKU caused by a defective PAH gene on chromosome 12

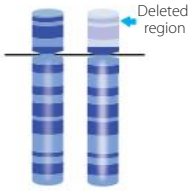


Chromosome 12

Chromosomal abnormalities

- Incorrect number of chromosomes
- Trisomy – one extra chromosome (e.g. Down syndrome – extra chromosome 21)
- Monosomy – one less chromosome
- A change to a chromosome – addition, substitution, deletion (e.g. cri du chat)
- Non-disjunction – incorrect separation of chromosomes during cell division





Chromosome 5 pair

Diseases caused by environmental exposure

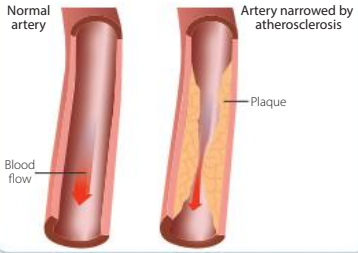
Lifestyle diseases

Risk factors include:

- tobacco use
- unhealthy diet
- physical inactivity
- harmful use of alcohol

Examples:

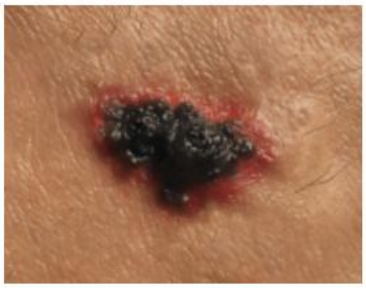
- cardiovascular disease, diabetes, cancers, chronic lung disease



Physical factors

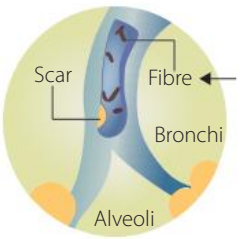
Exposure to physical factors in the environment

- e.g. exposure to UV light can cause skin cancer



Exposure to chemicals

- The type of disease depends on the chemical and the period of exposure
- Chemicals in land, waterways air, paint
- e.g. asbestosis caused by exposure to asbestos fibres



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Nutritional diseases

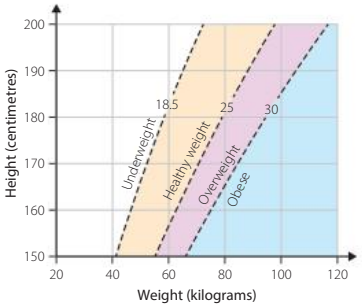
Caused by diet lacking balance and correct amounts of nutrients. This leads to *malnutrition*.

Undernutrition

- Insufficient intake of correct type or quantity of food, or both.
- Lack of protein – kwashiorkor
- Lack of energy and protein – Marasmus
- Vitamin deficiency:
  - vitamin A – blindness, dry skin
  - vitamin C – scurvy
- Mineral deficiency:
  - iron – anaemia
  - iodine – goitre
- Eating disorders
  - e.g. anorexia nervosa, bulimia nervosa

Overnutrition

- Consuming more kilojoules of energy than the energy expended leads to obesity
- Obesity leads to a number of health problems
  - e.g. high blood pressure, stroke, diabetes, cancer, reduced life expectancy



Cancer

- Abnormal cells divide in an uncontrolled way
- Caused by exposure to one or more risk factors
- Normal cell growth and mitosis is disrupted when regulatory genes (DNA repair genes, proto-oncogenes and tumour suppressor genes) are damaged or mutated, leading to uncontrolled cell replication, causing the formation of *tumours*.

Benign tumour

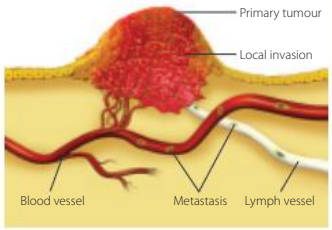
Not cancer – cells do not spread (metastasise).

Types of cancer

- Sarcoma – connective tissue
- Carcinoma – epithelial tissue
- Lymphoma – lymphatic system
- Leukaemia – blood marrow
- Central nervous system – brain and spinal cord

Malignant tumour

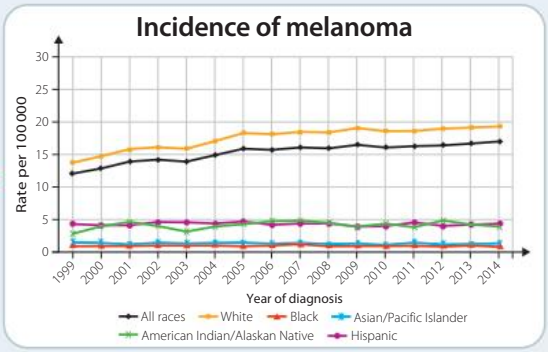
- Cells invade surrounding tissue
- Metastasis occurs
- New blood vessels develop to supply nutrients to tumour



Collecting and representing data

Incidence and mortality can be expressed as a rate per 100 000 population.

- Incidence – number of new cases in a specific time
- Mortality – number of deaths in a given time period
- Prevalence – number of people diagnosed who a disease that are still alive at the end of a given period



Prevalence of melanoma in the Australian population

YEAR DIAGNOSED	NO. PEOPLE LIVING AT THE END OF 2012 WHO HAD PREVIOUSLY BEEN DIAGNOSED WITH MELANOMA
2012	11 960
2008–2012	51 697
1982–2012	169 292

Source of data: Australian Government, Cancer Australia, 2018, 'Melanoma of the skin statistics', https://melanoma.canceraustralia.gov.au/statistics

15 CHAPTER REVIEW QUESTIONS

Review quiz

1 **a** What factors contribute to the development of non-infectious disease?

b Describe, using examples, why many non-infectious diseases are said to be preventable.

2 Refer to Table 15.1 on page 506. Assume that all the different types of cancer listed in the table were put into one group, called cancer.

a Calculate the:

i total number of deaths from cancer

ii percentage deaths from cancer.

b Where would this place cancer in terms of the top 10 causes of death in Australia in 2015?

3 Draw a flow chart summary to demonstrate how a mutation of a gene can cause disruption to a chemical reaction that is part of the metabolism.

4 **a** Identify the gene that is mutated in cystic fibrosis and the effects that this mutated gene has on the body of someone who has this disease.

b Describe the pattern of recessive inheritance that leads to inherited diseases such as cystic fibrosis occurring in the offspring of two parents who do not have the disease.

5 Explain why sufferers of phenylketonuria (PKU) must follow a low-protein diet for the rest of their lives.

6 Sufferers of Patau syndrome have developmental delays, small eyes, cleft lip and palate, organs that haven't developed properly and extra fingers and/or toes, among many other symptoms. Trisomy 13 causes this syndrome.

Explain what 'trisomy 13' means and how this could interfere with the proper functioning of the body.

7 **a** Identify the main risk factors associated with lifestyle diseases.

b Draw a flow chart to summarise the processes that occur in the disease atherosclerosis that lead to a cardiac arrest or a stroke.

c Distinguish between the terms 'ischaemic heart disease' and 'cerebrovascular disease'.

8 **a** Outline why prolonged exposure to heavy metals can cause disease in the body.

b Research the effect of prolonged exposure to mercury.

9 **a** Using the data in Table 15.11, draw a line graph to demonstrate the relationship between the age of the mother and the incidence of Down syndrome.

b Describe the trend shown in the graph.

c Give some reasons why this trend may occur.

10 A patient visiting a doctor presented with the following symptoms: reduced metabolic rate, low body temperature, lethargy, a visible lump in the throat region.

The doctor diagnosed a mineral deficiency.

a What was the mineral that was deficient and causing these symptoms?

b Research the foods that could be included in the patient's diet to increase the level of this mineral in the patient's body.

11 Identify and describe a nutritional disease that has a psychological basis, resulting in behavioural disorders that lead to an inappropriate diet and severe undernutrition.

12 Use the information about the BRCA1 gene on page 520 to answer the following questions.

a Outline the function of:

i a tumour suppressor gene

ii the BRCA1 gene

iii the PTEN gene.

b Draw a flow diagram that shows the links between the normal expression of the BRCA1 gene when there is damage to the PTEN gene and the maintenance and repair of body tissues.

c Draw a similar flow diagram to show the changes to this process as a result of a mutation to the BRCA1 gene.

13 Discuss how both environmental and nutritional factors could be risk factors that contribute to the development of cancer.

14 Prepare an answer to the inquiry question: 'Do non-infectious diseases cause more deaths than infectious diseases?'

TABLE 15.11 Effect of the age of the mother on the incidence of Down syndrome

AGE OF MOTHER	INCIDENCE (PER 1000 BIRTHS)
25	0.8
30	1.0
35	3.0
40	10.0
45	30.0
50	80.0

Exam preparation

9780170485067

16 Epidemiology

INQUIRY QUESTION

Why are epidemiological studies used?

Students:

- analyse patterns of non-infectious diseases in populations, including their incidence and prevalence, including but not limited to: ICT, IU, L, CC
  - nutritional diseases
  - diseases caused by environmental exposure
- investigate the treatment/management, and possible future directions for further research, of a non-infectious disease using an example from one of the non-infectious diseases categories listed above ICT, IU, L
- evaluate the method used in an example of an epidemiological study
- evaluate, using examples, the benefits of engaging in an epidemiological study

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
- **16.1** A secondary-source investigation to analyse patterns of a nutritional non-infectious disease in populations
- **16.2** A secondary-source investigation to analyse data about the incidence, mortality and prevalence of colorectal cancer
- **16.3** A secondary-source investigation of the treatment, management and future directions for a non-infectious disease
- **16.4** A secondary-source evaluation of the methodology of an epidemiological study
- **16.5** A secondary-source investigation to evaluate the benefits of an epidemiological study

Worksheets
- The parameters of epidemiology
- Types of epidemiological studies
- Comparing random and systematic errors
- Epidemiological studies crossword puzzle

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

The incidence of a particular disease, the cause of which is unknown, could be increasing in a population. This disease might be causing severe symptoms for the sufferers, and could quite often result in death. This leads scientists to ask questions such as: Are there patterns to this disease? What could be the cause of this disease? What control measures can be put in place to help prevent this disease?

In order to answer these questions, an epidemiological study can be undertaken. The word 'epidemiology' comes from *epi* and *demos*, Greek words meaning 'upon' and 'people or populace' respectively. It is a study of 'that which is upon people'. Epidemiologists study disease in a population, as opposed to those who study it in an individual organism. The World Health Organization's (WHO 2018) definition of epidemiology is:

> the study of the distribution and determinants of health-related states or events (including disease), and the application of this study to the control of diseases and other health problems. Various methods can be used to carry out epidemiological investigations: surveillance and descriptive studies can be used to study distribution; analytical studies are used to study determinants.
>
> Source: Word Health Organiation 2018, 'Health topics. Epidemiology', http://www.who.int/topics/epidemiology/en/

Dreamstime.com/Masterofall686

FIGURE 16.1 Epidemiology is the scientific study of patterns in data to help determine the cause of disease and the effectiveness of preventative strategies.

Epidemiology can be used to study both infectious and non-infectious diseases as well as events such as suicides, car accidents and work-related accidents. Epidemiology helps to determine what causes the disease, as well as who is affected, and where and how to intervene to prevent further occurrence. Studies can target limited public health resources at the specific causes and populations affected.

Public health authorities use the results of epidemiological studies to develop strategies for controlling disease and improving public health. Epidemiological studies can also be used to evaluate strategies that are already in place to control or treat disease and health-related conditions.

16.1 Analysis of patterns of non-infectious disease

Epidemiological studies play a major role in identifying patterns in the incidence, distribution, prevalence and mortality rates of disease. These studies also investigate the possible causes of disease and whether certain population groups are at greater risk of developing a certain disease. They go on to determine the strategies that would be most effective in controlling disease in the population.

Accepted scientific and mathematical models are used to statistically analyse the data that has been collected, to provide information about these trends. This analysis provides information to determine the trends for the overall population as well as for subsets of the population such as males, females, different races and different locations. Data is often presented in different forms, such as tables and graphs, to help understand the trends and patterns in data.

KEY CONCEPTS

- Epidemiology is the study of patterns of disease in populations.
- An epidemiological study can:
  - determine the cause of a disease and which populations are affected by the disease
  - help to develop strategies to control disease and improve public health
  - evaluate the effectiveness of strategies in place to treat/control disease.
- Analysis of data allows the identification of patterns and trends in the incidence, prevalence and mortality rates of disease.
- This information can then be used to identify areas and ways in which the overall health of the population can be improved.

Analysing data

Investigations 16.1 and 16.2 are about analysing patterns of non-infectious diseases in populations. In Investigation 16.1 you will analyse the data that you collected about a nutritional disease in Investigation 15.3 in the previous chapter. In Investigation 16.2 you will analyse the data presented about the incidence, mortality and prevalence of colorectal cancer in populations. It is your choice as to whether you complete only one of these investigations or both.

Worksheet
The parameters of epidemiology

INVESTIGATION 16.1

A secondary-source investigation to analyse patterns of a nutritional non-infectious disease in populations

INTRODUCTION

In this investigation you will analyse the data about a nutritional disease that you collected in the previous chapter. You will analyse the incidence and prevalence of the disease, although you may also wish to include analysis of other data such as mortality. Ensure that you use correct scientific terminology at all times.

Information and communication technology capability

Intercultural understanding

Literacy

Civics and citizenship

AIM

To analyse patterns of a nutritional disease in populations

METHOD

1. Review Chapter 15 and write down the definitions of incidence and prevalence (page 524).
2. For each set of data that you collected about your selected nutritional disease (in Investigation 15.3), describe the trend in the data for the incidence and prevalence of the disease, and describe any differences between the groups being studied.
3. Include analysis of data between different groups in the population such as gender, age, race and regions of the world.
4. If you have information in one particular form you could convert it into another form. For example, if you have tables containing data you could convert this information into graphs as part of the analysis.
5. Suggest reasons for each of the trends that you have described.
6. For at least one set of data, compose questions about trends in the data and use the data collected to answer the questions.
7. Predict future trends for each of the data sets you have studied.
8. Provide summary statements about the incidence, distribution and prevalence of your selected nutritional disease in Australia and the world. These statements should address the differences between groups in the population, based on age, gender, race and regions of the world.

»

» 9 Develop inquiry questions and hypotheses to identify a concept that could form the basis of future research into your chosen nutritional disease.

RESULTS

1 Include any graphs or tables that you have drawn from the data collected.

2 Record the questions that you have composed for one set of data and the answers obtained using that data.

DISCUSSION

1 Describe trends for each set of data collected about your selected nutritional disease and differences described between the groups being studied.

2 Suggest reasons for each of the trends you have described.

3 a Describe what data about prevalence indicates.

b Identify the link between prevalence data and the 5-year survival rate data.

4 Provide summary statements about the incidence, mortality and prevalence of your nutritional disease in Australia and the world. These statements should address the differences between groups in the population, such as age, gender, race and regions of the world.

5 Outline the inquiry questions and hypotheses you have developed to direct areas of future research into your chosen nutritional disease.

CONCLUSION

Write summary statements relating to the aim of this investigation.

INVESTIGATION 16.2

A secondary-source investigation to analyse data about the incidence, mortality and prevalence of colorectal cancer

INTRODUCTION

Information and communication technology capability

Intercultural understanding

Literacy

Civics and citizenship

Colorectal cancer, or bowel cancer, includes all cancers of the colon and the rectum. Of those people who develop colorectal cancer, 25% have some sort of hereditary influence. The other 75% of people who develop colorectal cancer have no family history of this cancer. Age is one of the risk factors associated with colorectal cancer, with a sharp increase in the incidence of colorectal cancer after the age of 50.

The chance of developing colorectal cancer is also increased by exposure to nutritional and environmental factors associated with lifestyle. Smoking and drinking alcohol are two of the lifestyle factors that increase the risk of developing colorectal cancer.

Nutritional behaviours such as eating red meat, especially when charred, and consuming processed meats that have been preserved, salted, cured or smoked, also increase the likelihood of developing colorectal cancer. Other risk factors, such as being overweight or obese and a lack of physical activity, also increase the chance of developing colorectal cancer.

AIM

To analyse patterns of colorectal cancer in populations

METHOD

1 Refer to the graph in Figure 16.2 of the estimated age-specific incidence and mortality rates for colorectal cancer in Australia, by sex, for 2017 and answer the questions in the *Analysis of results and discussion* section.

2 Refer to the graph in Figure 16.3 showing the age-standardised incidence and mortality rates for colorectal cancer in Australia and answer the questions in the *Analysis of results and discussion* section.

»

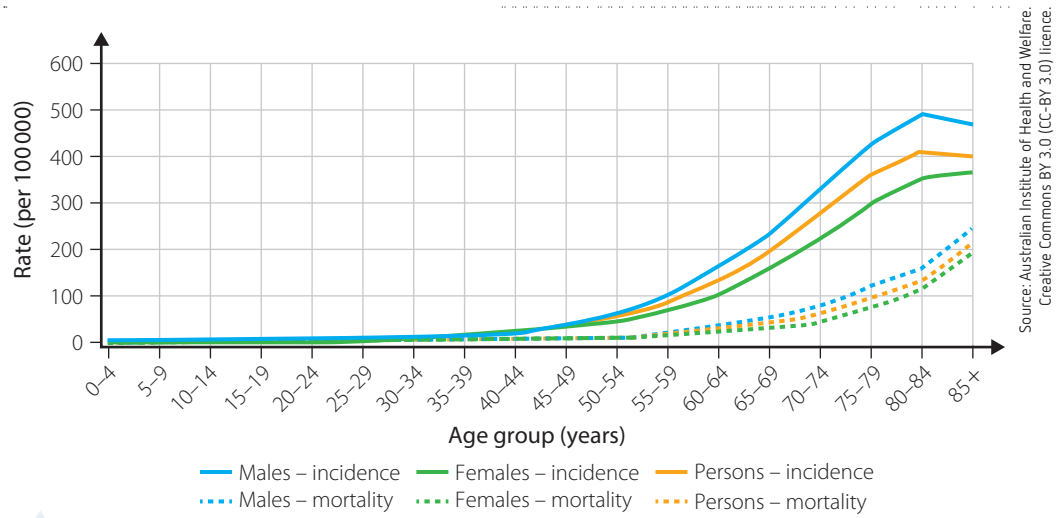


FIGURE 16.2 Estimated age-specific incidence and mortality rates for colorectal cancer, by sex, Australia 2017

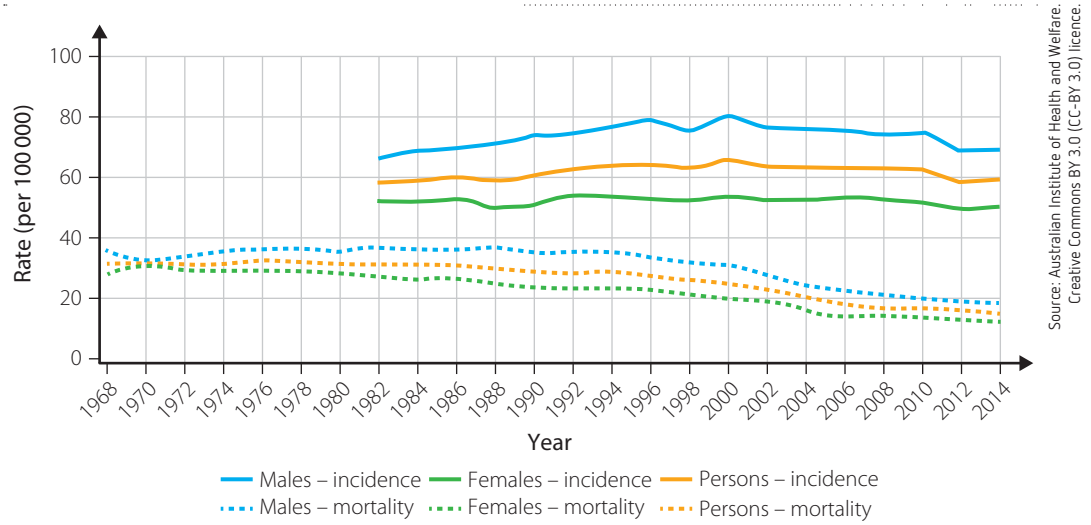


FIGURE 16.3 Age-standardised incidence rates for colorectal cancer 1982–2013 and age-standardised mortality rates for colorectal cancer 1968–2014, by sex, in Australia

3 Refer to the following information on the prevalence of colorectal cancer in Australia and summarise it in a table in the *Results* section.

At the end of 2012 there were:

- 13 078 people living who had been diagnosed with colorectal cancer that year
- 52 630 people who had been diagnosed with colorectal cancer in the previous 5 years (2008–2012)
- 129 497 people who had been diagnosed with colorectal cancer in the previous 31 years (1982–2012).

4 Refer to the graph in Figure 16.4, which shows the 5-year relative survival rate from colorectal cancer, 1984–1988 to 2009–2013, and answer the questions in the *Analysis of results* section.

The 5-year relative survival rate provides the percentage (%) chance of an individual being alive 5 years after being diagnosed with colorectal cancer.

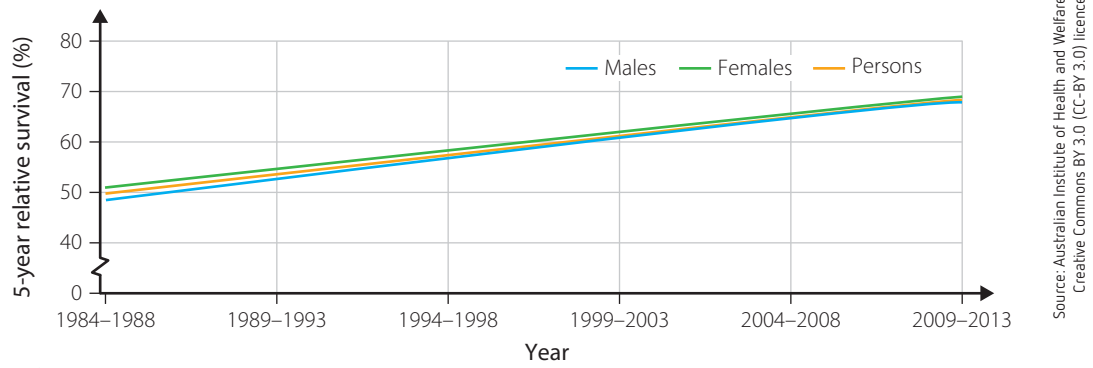


FIGURE 16.4 5-year relative survival from colorectal cancer, 1984–1988 to 2009–2013

5 Refer to Table 16.1, which lists incidence, mortality and prevalence data on colorectal cancer worldwide for 2012, and answer the questions in the *Analysis of results and discussion* section.

TABLE 16.1 Incidence, mortality and prevalence data worldwide for colorectal cancer, 2012

ESTIMATED NUMBERS (THOUSANDS)	MEN			WOMEN			BOTH SEXES		
	CASES	DEATHS	5-YEAR PREV.	CASES	DEATHS	5-YEAR PREV.	CASES	DEATHS	5-YEAR PREV.
World	746	374	1953	614	320	1590	1361	694	3544
More developed regions	399	175	1164	338	158	966	737	333	2130
Less developed regions	347	198	789	276	163	624	624	361	1414
WHO Africa region (AFRO)	16	11	32	15	11	31	31	22	63
WHO Americas region (PAHO)	125	57	362	121	55	342	246	112	705
WHO East Mediterranean region (EMRO)	18	12	40	15	10	33	33	21	73
WHO Europe region (EURO)	255	120	686	216	108	573	471	228	1258
WHO South-East Asia region (SEARO)	68	48	122	52	37	93	120	85	216
WHO Western Pacific region (WPRO)	264	125	711	195	100	518	460	225	1229
IARC membership (24 countries)	418	187	1181	351	167	976	769	353	2157
United States of America	69	29	214	65	27	199	134	55	413
China	147	79	338	107	60	245	253	139	583
India	37	28	50	27	21	37	64	49	87
European Union (EU-28)	193	83	536	152	69	417	345	152	953

Reproduced with permission from Ferlay J., Soerjomataram I., Ervik M., Dikshit R., Eser S., Mathers C., Rebelo M., Parkin D.M., Forman D., Bray, F. GLOBOCAN 2012 v1.0, Cancer Incidence and Mortality Worldwide: IARC CancerBase No. 11 [Internet]. Lyon, France: International Agency for Research on Cancer; 2013. Available from: http://globocan.iarc.fr, accessed on February 2017.

6 Refer to the graphs in Figure 16.5 showing the age-standardised incidence and mortality rate of colorectal cancer for all ages combined, by Indigenous status, in NSW, Vic., Qld, WA and NT. Then answer the questions in the *Analysis of results and discussion* section.

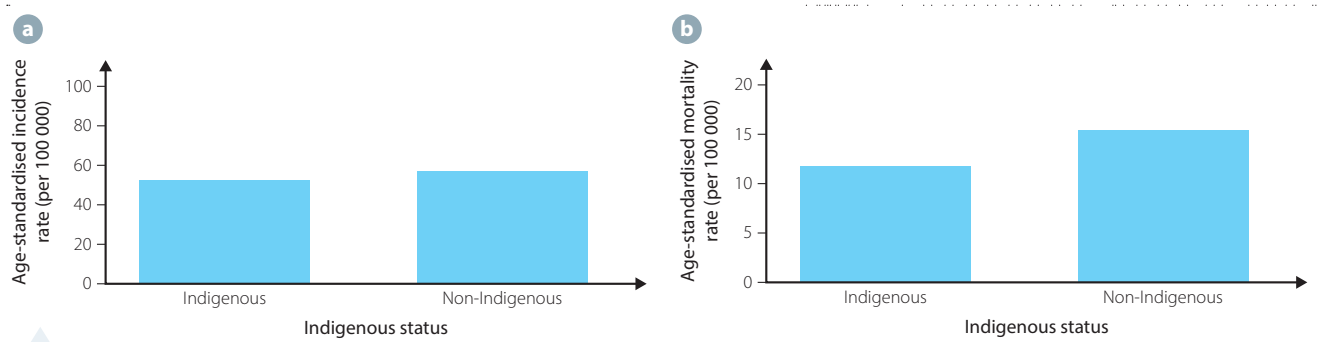


FIGURE 16.5 Colorectal cancer rates by Indigenous status: **a** age-standardised incidence rate of colorectal cancer for all ages combined, by Indigenous status (NSW, Vic., Qld, WA and NT, 2008–2012); **b** age-standardised mortality rate from colorectal cancer for all ages combined, by Indigenous status (NSW, Qld, WA, SA and NT, 2010–2014)

7 Refer to the graph in Figure 16.6 showing the age-standardised incidence and mortality rate for colorectal cancer in different regions of the world, and compose three questions relating to this data. Provide answers to these questions using the data.

For an explanation of the meaning of the term 'age-standardised' rates, see Chapter 15, page 524.

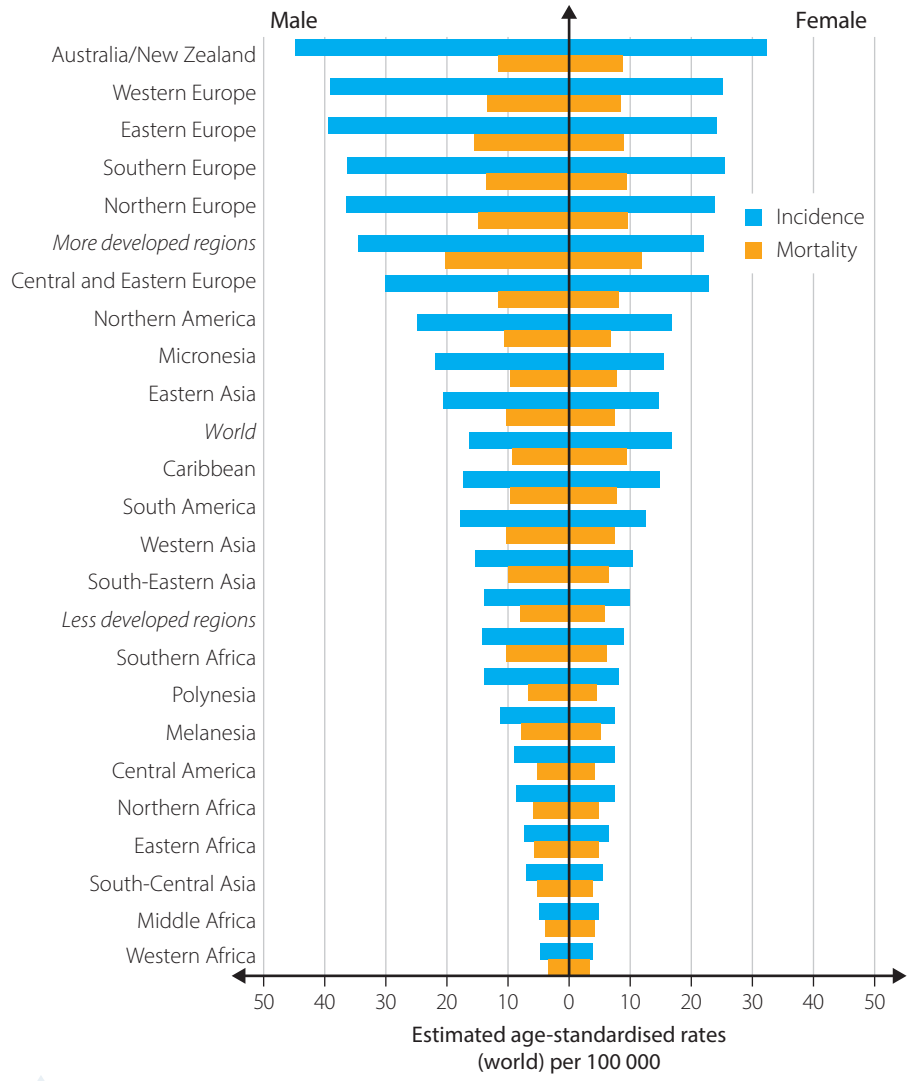


FIGURE 16.6 Age-standardised incidence and mortality rate for colorectal cancer in different regions of the world, 2013

RESULTS

1. Construct a table to summarise the information provided in Step 3 of the Method section.
2. Develop three questions about the data in Figure 16.6 and provide answers to these questions using the data.

ANALYSIS OF RESULTS AND DISCUSSION

When you are addressing each of the activities indicated, ensure that you use correct scientific terminology. Also ensure that you distinguish between measurements such as incidence (the actual number of new cases diagnosed) and the rate per 100 000 (either age-standardised or age-specific).

1. Refer to Figure 16.2. For both the age-specific incidence and mortality rates:
   - a Outline the trend shown for all persons.
   - b Outline the difference between males and females.
   - c Suggest hypotheses to account for the differences outlined.
   - d Predict future trends for each.
2. Refer to Figure 16.3. For both the age-standardised incidence and mortality rates:
   - a Outline the trend shown for all persons.
   - b Outline the difference between males and females.
   - c Suggest hypotheses to account for the differences outlined.
   - d Predict future trends for each.
3. Refer to Figure 16.3.
   - a Suggest reasons why both the age-standardised incidence and mortality rates have fallen in recent years.
   - b Predict future trends for each. Explain the basis of your prediction.
4. Refer to Figure 16.4, Table 16.1 and the table you constructed showing prevalence.
   - a Describe, in general terms, what the data about prevalence indicates.
   - b Identify the link between the prevalence data and the 5-year survival rate data.
   - c Outline the trend in the 5-year survival rate for colorectal cancer.
   - d Suggest reasons for the trend shown.
   - e Predict future trends in the 5-year survival rate for colorectal cancer in Australia.
   - f Compare the 5-year prevalence data for colorectal cancer in different regions of the world.
5. Refer to Figure 16.5.
   - a Comment on the difference in incidence and mortality rate for colorectal cancer between the Indigenous and non-Indigenous populations.
   - b Develop inquiry questions and hypotheses to investigate/research reasons for these differences.
6. Provide summary statements about the incidence, mortality and prevalence of colorectal cancer in Australia and the world. These statements should address the differences between different groups in the population such as those defined by age, gender, race and regions of the world.

Weblink
Incidence and prevalence of cancers
Watch the videos about different types of cancer to determine the incidence and prevalence of different types of cancer.

CONCLUSION

Write summary statements relating to the aim.

1 The graph in Figure 16.7 shows the age-standardised incidence rate for lung cancer, 1982–2013, and the age-standardised mortality rates for lung cancer, 1968–2014, by gender, in Australia.

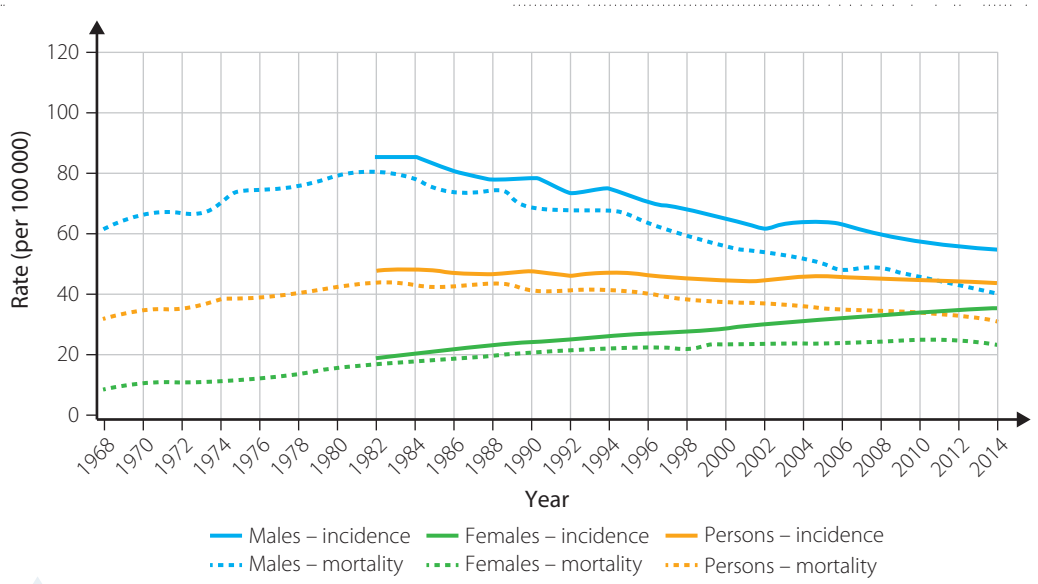


FIGURE 16.7 Age-standardised incidence rate for lung cancer, 1982–2013, and age-standardised mortality rates for lung cancer 1968–2014, by gender, in Australia

a Outline the trend shown in the graph for the age-standardised *incidence* rates of lung cancer for:
- **i** females
- **ii** males.

b Suggest hypotheses to account for these trends.

c Outline the trend shown in the graph for the age-standardised *mortality* rates of lung cancer for:
- **i** females
- **ii** males.

d Suggest hypotheses to account for these trends.

e Predict future trends in the age-standardised mortality rates of lung cancer in both males and females.

f Describe how this information could be used to improve the health of the population.

2 Refer to the graphs in Figure 16.8 showing the prevalence of diabetes mellitus (types 1 and 2) for different age groups in the Australian population in 2001 and 2014–2015. These graphs show the percentage (%) of the population in different age groups that had diabetes mellitus.

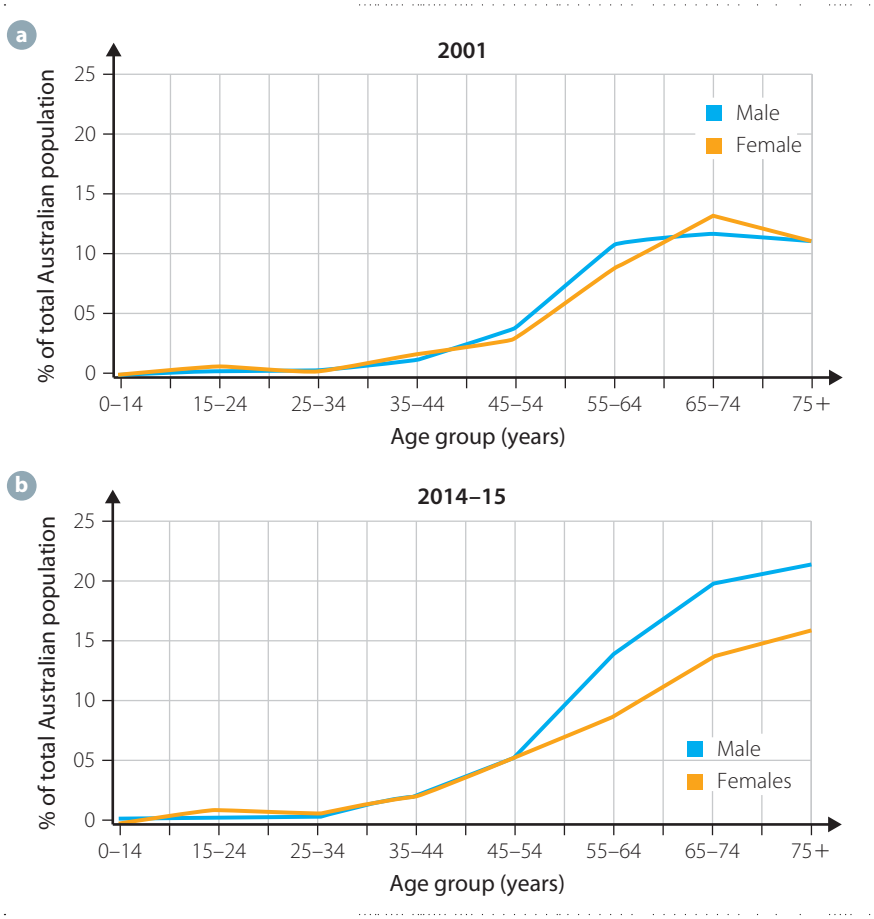


FIGURE 16.8 Prevalence of diabetes mellitus for different age groups as a percentage of the total Australian population, **a** in 2001 and **b** in 2014–2015

a For both 2001 and 2014–2015:

i Which age group for both males and females had the highest percentage of individuals with some form of diabetes?

ii Describe the overall trend for the percentage of the population with diabetes mellitus.

b **i** Fill in the following table with information obtained from the appropriate graph.

	PROPORTION OF THE POPULATION WITH DIABETES MELLITUS (%)			
	MALES		FEMALES	
YEAR	AGE 45–54	AGE 75+	AGE 45–54	AGE 75+
2001				
2014–2015				

ii Compare the proportion of males with diabetes between the age groups shown in 2001 and 2014–2015.

iii Compare the proportion of females with diabetes between the age groups shown in 2001 and 2014–2015.

iv Outline the trends shown in the proportion of males and females with diabetes mellitus in 2014–2015.

3 **a** Define 'epidemiology'.

b Outline why epidemiological studies are used.

16.2 Treatment, management and future directions

Treatment of a disease will depend on the type of disease. Some non-infectious diseases, such as vitamin or mineral deficiency, simply require the correct amount of the vitamin/mineral to be included in the diet, whether in food or by supplementation. The treatment of the nutritional disease scurvy, which is caused by lack of vitamin C, simply involves adding appropriate amounts of vitamin C to the diet either by eating foods rich in vitamin C or by taking vitamin C supplements.

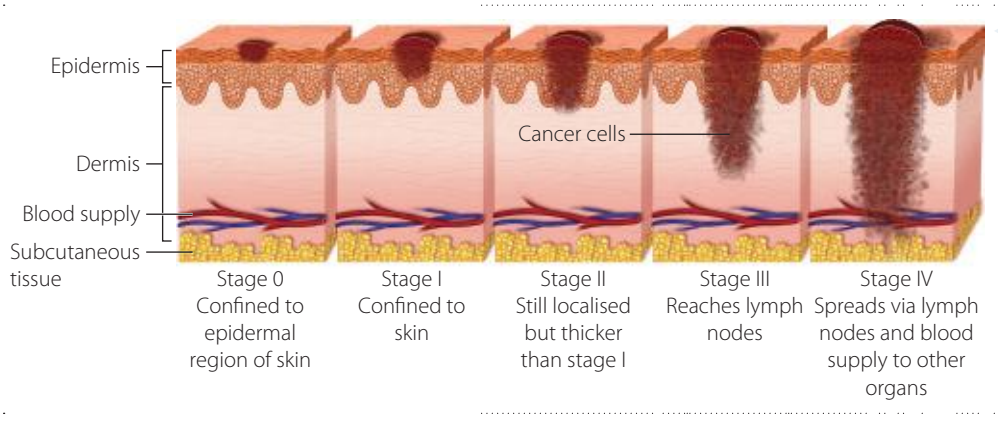
Other diseases, such as cancer, require more complex treatments to try to cure the individual.

Some diseases, especially those caused by genetic factors, have limited treatments available, and so screening, early diagnosis and management of the symptoms is the best way to deal with the disease.

Research into many types of diseases, a greater understanding of the way in which the disease manifests in the body, along with developments in the understanding of the way systems in the body work, all contribute to the development of more successful treatments for disease. For example, the disease cystic fibrosis has no successful treatment at present and management strategies concentrate on reducing the severity of the symptoms. Research is currently being undertaken into the use of gene therapy to treat this disease.

Melanoma: a disease caused by environmental exposure

There are many treatment options available to sufferers of melanoma, depending on the stage at which the disease is diagnosed (Fig. 16.9).



ilusmedical/Shutterstock.com

FIGURE 16.9 The stage at which melanoma is diagnosed determines the best treatment option.

Treatment

The most common treatment for melanoma that has been detected early is surgery, and this is often the only treatment required. Surgery involves removing the tumour and the skin around the tumour to ensure that none of the cancerous cells are left behind, to reduce the risk of a recurrence of the melanoma.

For more advanced cases of melanoma, a range of treatment options are available, including radiation, chemotherapy, targeted therapies and immunotherapy. The type of treatment used will depend on numerous factors, including how far the melanoma has progressed, and the person's age and health.

Radiation

When cells are exposed to X-rays, normal cells can repair their DNA more easily than cancer cells can. X-rays damage the DNA of the cancer cells and kill them. Care must be taken in directing the radiation to the specific cells that are intended for destruction. This radiation can be administered externally or by placing a radioactive source in the body near the cancer cells.

Chemotherapy

Chemotherapy drugs slow the growth of cancer cells, but they have not been particularly effective in treating melanoma, and so are not used to treat this form of cancer as much as in other cancers.

Weblink
Treatment for melanoma
Watch the video and summarise the main treatments for melanoma.

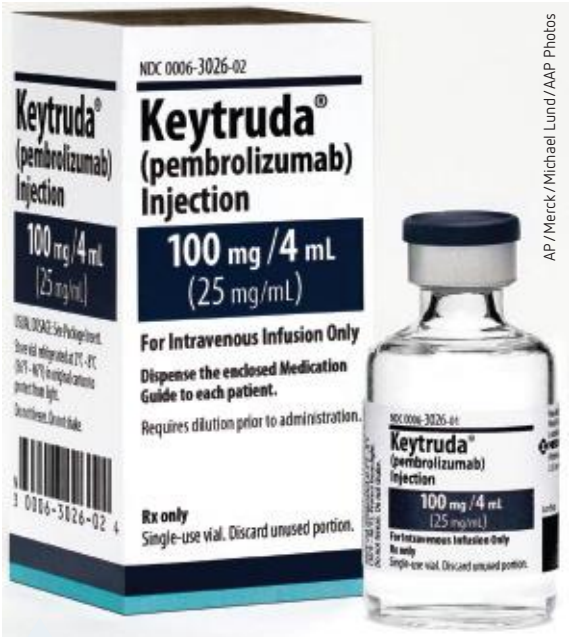


FIGURE 16.10 Immunotherapy drugs, such as Keytruda®, cause the immune system to recognise and destroy melanoma cells.

Targeted therapies

Targeted therapies involve the use of drugs that affect the molecules that control the growth of the tumour cells, which effectively stops the growth and spread of the tumour to other organs. Melanoma cells have mutations that cause the uncontrolled division of cells, leading to the growth of tumours. Targeted therapy drugs interrupt the pathways that cause this uncontrolled division of melanoma cells.

Immunotherapy

Immunotherapy treatments effectively cause the body's own immune system to fight the melanoma. Two approaches are in the early stages of development and use.

One of the approaches is to use drugs called '**checkpoint inhibitors**' that cause the immune system to recognise and destroy melanoma cells (Fig. 16.10). Cancer cells cause the immune system to ignore them. Checkpoint inhibitors reverse this effect and have proved successful in a number of patients.

The other approach is to use vaccinations as a method of treatment rather than prevention. An antigen is produced using the melanoma cells, and when injected allows the immune system to more easily identify and destroy the melanoma cells.

Future directions

Future research is required to further develop targeted therapies and immunotherapy treatments for melanoma. The use of targeted therapies is based on the different types of mutations that cause the uncontrolled cell division that occurs in melanoma. Drugs have been developed to interrupt these specific pathways. Scientists are aware that numerous other mutations are yet to be identified, and so research is continuing in order to identify these. Once these mutations are identified, further drugs will then be developed to interrupt many more of these pathways.

Currently there are only two immunotherapy drugs available for use and these are not successful on all patients with melanoma. Further research is required to develop a greater variety of immunotherapy drugs, so that the majority of patients can benefit from this form of treatment of melanoma.

The use of vaccines to treat melanoma is in its early stages. More research is required to further refine this process and make it consistently effective. Further research may also involve investigating the relationship between melanoma and other cancers.

INVESTIGATION 16.3

A secondary-source investigation of the treatment, management and future directions for a non-infectious disease

INTRODUCTION

In this investigation you will concentrate on a non-infectious disease selected from nutritional diseases or those caused by environmental exposure. Of the many types of these diseases, some can be treated successfully. For others, however, there is currently no successful treatment and so the only option is to manage the symptoms.

In diseases where treatment is either non-existent or not fully effective, research is an important tool to try to develop appropriate treatments to improve the health outcomes of those with the disease.

AIM

To outline the treatment/management of, and possible future directions for further research into, a nutritional disease or a disease caused by environmental exposure

Information and communication technology capability

Literacy

METHOD

1. Choose a nutritional disease or a disease caused by environmental exposure. Review the information in Chapter 15 about these types of diseases.
2. Research, using a wide range of reliable secondary sources, to find the following information about your selected disease:
   - **a** current methods of treatment and/or management
   - **b** possible future directions for research to improve treatment and/or management.
3. You may wish to add to your knowledge of the treatment/management by talking to a person who has the disease, or to a doctor.

   Before you conduct the interview, you should carefully formulate the questions you are going to ask, in order to obtain the information you require.

   Research ethical approaches and questions to ask, and questions to avoid asking in order not to offend the person being interviewed.
4. Present your information in a style of your choosing that is clear, easy to read and consistent with the type of information you collect. Ensure that you use appropriate scientific language.
5. Assess the accuracy, reliability, validity and relevance of all data obtained.
6. Acknowledge your sources, using an accepted referencing style.

RESULTS

1. Present information about your selected disease in regards to:
   - **a** treatment and management of the disease
   - **b** possible future directions for further research to improve the treatment and/or management of this disease.
2. Include in your presentation an assessment of the accuracy, reliability, validity and relevance of all data obtained.

Refer to Chapter 1, page 10, for guidance on how to assess the relevance, accuracy, validity and reliability of sources.

DISCUSSION

1. Distinguish between the treatment and management of a disease.
2. Discuss the importance of treatment/management of non-infectious disease.

CONCLUSION

Write summary sentences that address the aim of this investigation.

KEY CONCEPTS

- Treatment of non-infectious disease depends on the type of disease.
- Management of symptoms is the only option for non-infectious diseases that have no successful treatment strategies.
- Treatment options for melanoma include surgery, radiation, chemotherapy, targeted therapies and immunotherapy.
- Further research is required to develop a greater variety of targeted therapy and immunotherapy drugs and vaccines to treat melanoma.

CHECK YOUR UNDERSTANDING 16.2

1. Distinguish between the terms 'treatment' and 'management' when referring to non-infectious disease.
2. List, in order of effectiveness from least to best, the options available to treat melanoma.
3. Scientists are concentrating on uncovering the mutations that occur in skin cells as part of their quest to develop future treatments for melanoma. Explain why they are doing this.

16.3 Methods used in epidemiological studies

There are three major types of epidemiological studies:

- descriptive
- analytical
- intervention.

Descriptive and analytical studies are **observational studies**, which help us to understand the causes of diseases.

Descriptive studies

Descriptive studies are usually the first type of study conducted when investigating the cause of a disease. These studies provide information about the patterns of the disease, including the frequency of the disease, which section of the population is affected (age, gender, occupation, socioeconomic status and so on), the geographical location and whether there was a particular time period in which individuals were affected. Data including information about age, sex, diet, occupation, drinking habits, location of work and home and places visited are collected from individuals with the disease, and commonalities are determined in order to find a possible cause. Hypotheses are proposed about the cause of the disease.

In an early epidemiological study undertaken to determine the cause of lung cancer, the data collected included, among other things, information about the age, sex, smoking habits, diet, occupation and drinking habits of people with lung cancer and people without lung cancer.

Analytical studies

Once a descriptive study has been completed, **analytical studies** are used to collect more data, which is then statistically analysed to test hypotheses as to the likely cause(s) of the disease. The *morbidity* (number of cases of the disease) and the *mortality* (percentage of the population that dies from the disease) are two indicators that can be used in these studies. Data about the *incidence* (number of new cases in a specific period) and the *prevalence* (number of people affected at any one time) are also compiled in these studies.

Case-control studies and cohort studies are two types of analytical studies that can be used.

Case-control studies

Case-control studies compare people with the disease (cases) to people without the disease (controls) and look for differences in exposure to the possible causes of the disease (Fig. 16.11). A large range of data, including age, sex, diet, location, lifestyle, occupation and exercise habits is collected from both groups and then analysed to determine the likely cause of the disease.

A case-control study that was set up in London in 1947 by Richard Doll compared patients with lung cancer to patients with other conditions. Information about many of the factors in their lives, including their smoking habits, was collected and analysed. The results of this study showed that most of the individuals with lung cancer were smokers. Doll was the first to suggest a link between smoking and lung cancer.

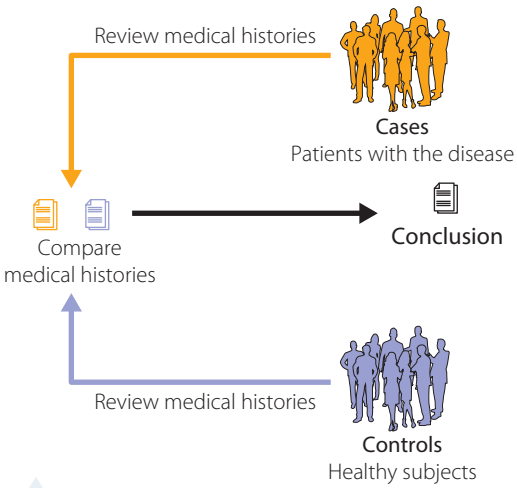


FIGURE 16.11 The principles of a case-control study

Cohort studies

Cohort studies involve studying two or more similar groups of people who are free of the disease (Fig. 16.12). These groups differ in one main factor: their exposure to the potential cause of the disease. One of the groups is exposed to the possible cause of the disease and the other group is not. These groups are followed over a long period of time, to compare the resulting incidence of the disease being studied.

For example, after the 1947 case-control study that established a link between smoking and lung cancer, A. B. Hill conducted a cohort study in England in 1951. This study followed more than 40 000 doctors over a 10-year period. One group of doctors were smokers (the test group) and the other group were non-smokers (the control group). At the end of the study it was found that the test group had a much higher incidence of lung cancer than the control group. This study also revealed that the greater the number of cigarettes smoked daily, the greater the chance of developing lung cancer.

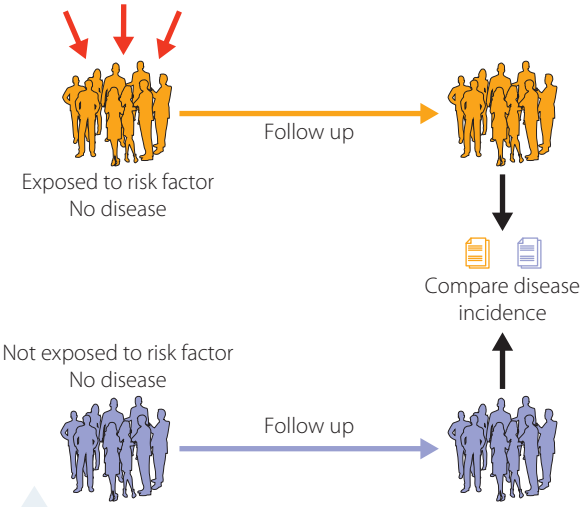


FIGURE 16.12 The principles of a cohort study

Intervention studies

Intervention studies are used to test the effectiveness of a treatment (for example, a clinical trial of a new drug) or the effectiveness of a public health campaign. The aim of an intervention treatment is to change the behaviour of the population as a whole in order to reduce the incidence of the disease.

One type of intervention study is an **experimental study**, which is often used to test the effectiveness of a new type of drug. In this type of study, people who are suffering from a particular condition are observed for a set time period. Participants are randomly placed into two groups. One group receives the trial drug, while the other group receives a **placebo**. The effects of the 'medication' on individuals in each group are recorded and statistically analysed to determine the effectiveness of the drug being studied.

If it is impossible to set up a randomised trial, a **quasi-experimental study** is carried out. This differs from an experimental study in that the researcher chooses the subjects who receive the drug/treatment.

Weblink
Quit now
Read the methodology, effectiveness and conclusions drawn from this campaign.

An example of this is the testing of the effectiveness of a vaccine for influenza in hospital workers. Workers in one department of the hospital would be given the vaccine and those in another department would not – the incidence of influenza would then be compared between the two groups. In a further example, the effectiveness of campaigns such as the 'Quit now – smoking while pregnant' campaign to reduce the number of pregnant women smoking can be evaluated using an intervention study.

The types of epidemiological studies are summarised in Figure 16.13. Epidemiological studies should:

- be conducted over a long period of time
- study very large sample sizes (thousands)
- collect a range of relevant data from a large group of both affected and unaffected people (case-control). This data could include age, sex, diet, occupation, lifestyle and exercise habits
- have included participants that represent the wider population
- use control groups who are not exposed to the potential cause of disease but are similar in all other respects to the test group (cohort studies)
- collect data on the incidence, prevalence, mortality and morbidity rates of the disease being studied
- statistically analyse the data to identify patterns and trends in the occurrence of the disease
- identify the possible cause of the disease and any risk factors
- develop a management plan with strategies to control or eliminate the disease and educate the public
- evaluate the effectiveness of control and treatment programs.

Worksheet
Types of epidemiological studies

Weblink
Disease detective
Work through this interactive activity as an epidemiologist trying to determine the cause of a disease.

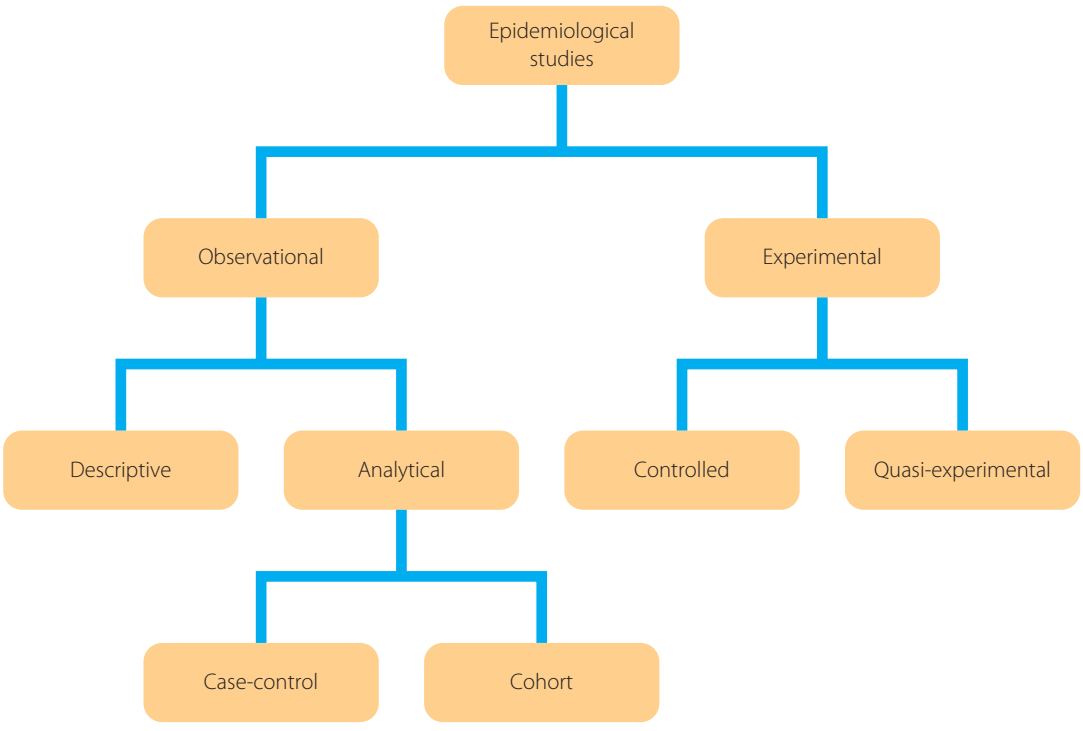


FIGURE 16.13 The different types of epidemiological studies

Errors in epidemiological studies

There are two major types of errors in epidemiological studies:

- random errors
- systematic errors.

Random errors

Random errors are unpredictable variations in the data and have an inconsistent effect on measurement within a study. They make the study less precise but do not shift the results of the study in a particular direction. The effects of random error can usually be corrected using statistics. Random errors can occur simply through differences in the subjects being studied. Therefore, ensuring that the separate groups being studied are homogenous will increase the precision of the study and reduce random error. Ensuring a large sample size for the study can also reduce random error.

Systematic errors

Another name for **systematic errors** is **bias**. This encompasses any process during the study that causes a consistent deviation in measurement from what the true value should be. Systematic errors result in an incorrect estimate of the effect of exposure to a particular factor and the cause of a disease.

Revise random and systematic errors in measurement in Chapter 1, pages 19–21.

The two main types of bias are selection bias and information bias.

- **Selection bias** is bias in selecting subjects to include in the study. In any study, the subjects must be representative of the population that is to be studied. A number of factors in the selection process can cause bias, so that the sample studied is not representative of the population. These factors include:
  - **sampling bias**, in which the way the subjects are chosen or where they are chosen from does not lead to a sample that represents the population that is to be studied
  - **volunteer bias**, in which those who volunteer for a study have a vested interest in the condition in some way and may already be at a higher risk than those who do not volunteer
  - **healthy worker bias**, in which participants who are working (in employment) are generally healthier than participants who are not. If only working participants are included in the study, then those who have left employment due to ill health will not contribute, as they should, to the study, especially if the study is related to exposure at the place of employment
  - **prevalence/incidence bias**, in which only current cases are included in the study. Those who have recovered or died should also be included.
- **Information bias** involves errors in taking measurements or recording information. Inaccurate or incomplete measurements and observations will result in information bias if the inaccuracy affects each study group differently. Some of the different types of information bias are:
  - **misclassification bias**, in which some of the subjects are already suffering from the condition and are undiagnosed at the start of the study
  - **recall bias**, in which subjects' ability to recall varies. Those who are affected by the condition will have a much greater recall of factors than those who have not been affected
  - **ascertainment bias**, in which all members of the study group are not followed up to the same degree
  - **interviewer bias**, in which the interviewer indirectly leads the study participant to the answer to the questions
  - **measurement bias**, in which measurements are consistently inaccurate (overstated or understated). This can be reduced by using multiple observers, taking repeated measurements, and establishing standards and objective definitions of measurements beforehand
  - **loss to follow-up bias**, in which not all subjects who began the study are available at the end of the study.

Worksheet
Comparing random and systematic errors

A **confounding factor** is a type of systematic error that occurs when an unrecognised factor may be affecting the results of a study and leading to bias. A particular factor may be attributed as having caused a disease, but another factor could also have contributed to the same disease. For example, asbestos workers and non-asbestos workers were studied to find the occurrence of lung cancer in each group. A confounding factor that needed to be taken into account in this study is whether the study participants were smokers, as this factor could also contribute to the development of lung cancer.

KEY CONCEPTS

- Large sample size and long periods of study are important requirements of epidemiological studies.
- Descriptive, analytical and intervention studies are the major types of epidemiological studies.
- Descriptive studies provide the *who*, *what*, *where* and *when*, and generate hypotheses about causes of disease.
- Analytical studies test hypotheses and provide the *why* and *how*, to determine the cause of the disease.
- Intervention studies are used to test the effectiveness of a treatment for a disease, or the effectiveness of a public health campaign.
- Random and systematic errors can occur in epidemiological studies.
- Random errors reduce precision but do not skew (bias) the results of a study.
- Systematic errors shift the results of a study in a particular direction and include selection bias, measurement bias and confounding factors.

CHECK YOUR UNDERSTANDING 16.3a

1. Construct a table to summarise the different types of epidemiological studies, the methods used in each and the types of information obtained.
2. An epidemiological study was carried out to identify the risk factors that could lead to obesity. In this study, thousands of obese individuals from a wide range of locations and societies were surveyed about a large number of factors in their life, including their diet, level of activity, occupation, amount of sleep, socioeconomic status, gender, hair, skin and eye colour, and ethnic background.
   a. Identify the type of epidemiological study carried out in the example above.
   b. What would be the purpose of this study?
   c. Describe further investigations that could be undertaken, to obtain more data about obesity.
   d. Outline how health authorities would use the data obtained from the initial and subsequent studies.
   e. Outline the two types of errors in epidemiological studies.
3. Construct a table to summarise the types of systematic errors.

Evaluating an epidemiological study

Intercultural understanding

Epidemiological study: The Pima Indian population

Many independent studies have linked increased physical activity with a change in body mass and composition, with all these factors contributing to a reduced incidence of type 2 diabetes. A cohort study was carried out on the Pima Indian population, in the Gila River Indian Community, a native American population living in Arizona (Fig. 16.14). This study examined the role of physical activity in the development of type 2 diabetes (independently of the resulting change to body mass and composition). This study was carried out between 1987 and 2000 with 1728 non-diabetic Pima individuals aged 15–59 years.

The Pima Indian population has been part of a longitudinal population-based study of diabetes since 1965. In this study, anyone who lives in a designated part of the community and is over the age of 5 years can participate. As part of this study, the same individuals are examined every two years and a glucose test is performed to determine whether the person has type 2 diabetes. A physical examination is also performed, along with measurement of height and weight and the recording of the person's medical history.

Method

- From 1987, as well as the usual testing and measurement, the participants in the study who were 15–59 years old were also interviewed about their physical activity. The interview was conducted by trained personnel who implemented a scientifically valid and reliable questionnaire.
- Each participant's leisure and occupational activity for the past year was assessed separately. Only physical activity that was greater than normal everyday tasks such as bathing and dressing was used.
- From a list of common activities, participants were asked to report on their leisure activities over the past year, along with the frequency and duration of each activity.
- To measure occupational activity, the participants were asked to report on all the jobs they had had in the previous twelve months. For each job, the average work schedule (months, days and hours), time spent walking or riding to work, the number of hours spent sitting and the most common activities that were performed when not sitting, were recorded.
- Activities were weighted according to their intensity, using scientifically accepted models. Estimates were calculated separately for both the leisure and occupational activities as hours per week and averaged over the previous year. The activity level of each individual was subsequently classed as either high or low.
- If the interviewer judged that a participant was incapable of reporting their activity correctly, their results were excluded from the analysis.
- To be included in the analysis, participants had to have a baseline measure of activity and at least one follow-up visit. Values for each individual were calculated from the baseline measurements until diabetes developed or until the last examination.
- Incidence rates were calculated by age group, gender and physical activity using a scientifically approved model.

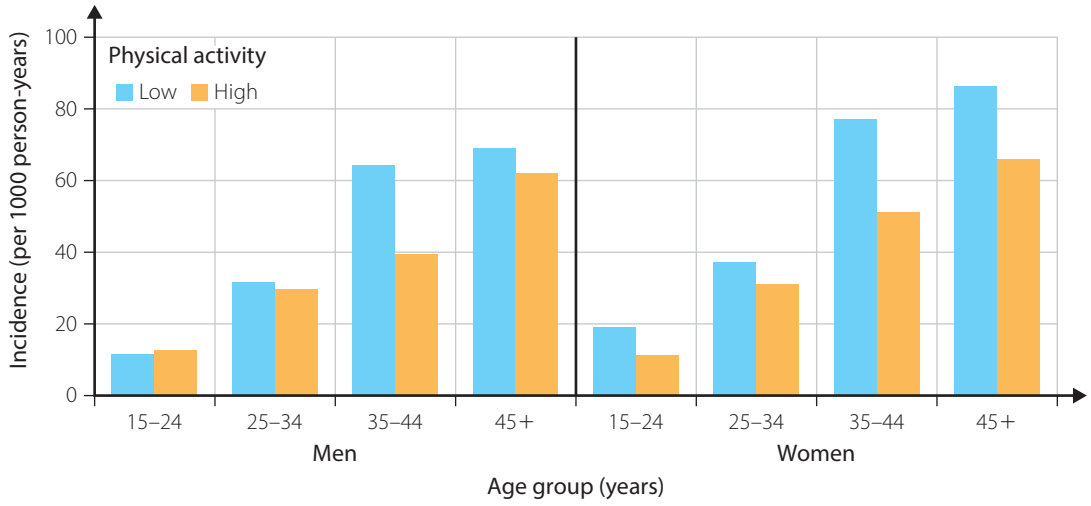
The results of this study indicated that, in most age groups, and for males and females, the incidence rate of diabetes was lower in those who had higher levels of physical activity than in those who had low levels of physical activity (Fig. 16.15).

Weblink
Pima Indians
Watch the video to determine the prevalence of diabetes in the Pima population.

David Ryan / Alamy Stock Photo

FIGURE 16.14 Pima Indians from the Gila River Indian Community

FIGURE 16.15
Diabetes incidence rates by age and gender for total physical activity levels, from the study of 1728 Pima Indians without diabetes, Gila River Indian Community, 1987–2000



Source: American Journal of Epidemiology, Physical Activity, Obesity, and the Incidence of Type 2 Diabetes in a High-Risk Population Andrea M. Kriska Aramesh Saremi Robert L. Hanson Peter H. Bennett Sayuko Kobes Desmond E. Williams William C. Knowler American Journal of Epidemiology, Volume 158, Issue 7, 1 October 2003, Pages 669–675.

Evaluation

To *evaluate* involves making a judgement about something and using evidence to support your judgement. The evidence used should be based on specific criteria relevant to the particular scenario being evaluated.

The validity of the method used in an epidemiological study should be evaluated on whether it follows accepted epidemiological principles for the particular type of study being carried out. Any errors and bias, including **confounding variables**, should be considered when assessing the validity of the study.

Criteria common to most epidemiological studies include large sample size, long period of study, use of scientifically approved methods of conducting the study, collecting data and analysing results. Other criteria will depend on the type of study – for example, whether it is a case-control, cohort or experimental study.

In the Pima Indian example:

- The study size of 1728 individuals and the 13-year time period over which it occurred satisfy the epidemiological requirements of a large sample size and a long period of study, reducing the effect of sampling bias.
- In a cohort study such as this, two or more similar groups of people who are free of the disease should be studied. The major difference between the groups should be the factor that is being studied. This study satisfies these requirements, as non-diabetic Pima Indians from the same designated areas and aged 15–59 were studied. The groups varied in the amount of physical activity that was part of their daily life.

Other factors also ensured the validity of the Pima Indian study:

- The diagnosis of diabetes was made by objective means – scientifically approved testing at the first and each follow-up visit. This reduced the likelihood of measurement bias.
- Trained interviewers used a scientifically valid questionnaire to determine the activity levels of participants. The trained interviewers and objective questionnaire reduced both interviewer and measurement bias.
- Using mathematical models, participants' physical activities were weighted for their intensity level, and the activity levels of each individual were then classified as either high or low.
- Scientifically tested models were used to analyse the results and determine the incidence rates of diabetes related to activity levels and BMI index.
- Data was excluded from analysis if individuals were thought to have incorrectly reported activity levels, which reduced recall bias.
- The written report of the study was peer reviewed before publication.

The methodology used in this epidemiological study is judged to be valid, as it follows the epidemiological principles outlined earlier.

Worksheet
Epidemiological studies crossword puzzle

9780170485067

INVESTIGATION 16.4

A secondary-source evaluation of the methodology of an epidemiological study

INTRODUCTION

In this secondary-source investigation you will research secondary sources to find an example of an epidemiological study and evaluate the method used in this study. Working in pairs or groups would be beneficial in the successful implementation of this investigation. A suggestion is that you prepare and present a report in an appropriate way to the class for peer review.

AIM

To evaluate the validity of the method used in a sourced epidemiological study of a non-infectious disease/condition or related component

METHOD

1. Either individually, or in pairs or groups, use a variety of secondary sources to research an epidemiological study about a topic of interest.
2. Some options you may wish to research:
   - an experimental study such as a randomised controlled trial of a new drug or treatment for a non-infectious disease/condition
   - a case-control study aimed at determining the cause(s) of a non-infectious disease
   - a cohort study aimed at identifying factors that affect the incidence of a particular non-infectious disease.
3. Identify the type of epidemiological study you have chosen and make a summary of the method used.
4. Choose the criteria that you are going to use to make a judgement about the validity of the method. This will depend to some extent on the type of study you have chosen.
5. In a table, list the criteria to be used and the components of the method used in the study that satisfy these criteria.
6. Provide a bullet point summary of any limitations identified in the methodology of your chosen study. This should include any evidence of bias in the study.
7. Make a judgement about the validity of the method used in the study and provide a summary of the evidence that supports your judgement.
8. Prepare a scientific argument that supports or refutes the claims made in the study based on your evaluation of the method.
9. Present your information in an appropriate manner, either for a written report or a class presentation for peer review if required.
10. Reference your researched information in an acceptable way.

For acceptable methods of scientific referencing, refer to Chapter 1, page 27.

RESULTS

1. Provide a summary of your chosen epidemiological study.
2. Construct a table, such as the one below, to match the requirements of a valid method in epidemiological study with the components of your chosen study.

CRITERIA FOR VALID METHODOLOGY	COMPONENTS OF CHOSEN STUDY

3. Make a bullet point summary of any limitations identified in the methodology of your chosen study.
4. Present your judgement about the method used and a summary of evidence, in an appropriate way.
5. Prepare an appropriate class presentation, if required.

»

» DISCUSSION

1 Outline the process to follow when you are asked to 'evaluate' something.
2 Discuss ways in which identified limitations in the method of your chosen study could be improved.
3 Outline the benefits of working in a team.
4 Assess the relevance, reliability, validity and accuracy of the secondary sources used in this investigation.

CONCLUSION

Provide summary sentences that address the aim of this investigation.

KEY CONCEPTS

- To 'evaluate' means to make a judgement about something, based on certain criteria. Evidence should be provided to support the judgement that has been made.
- Evaluation of the validity of the method used in an epidemiological study is determined by how well it follows accepted epidemiological principles.
- Along with large sample size and long periods of study, the use of scientifically approved methods of implementing a study, collecting data and analysis of results are the main requirements of epidemiological studies.

CHECK YOUR UNDERSTANDING 16.3b

1 State the hypothesis being tested in the Gila River Indian Community study.
2 How was the study sample determined?
3 What quantitative measurements were taken of this sample?
4 What qualitative measurements were taken of this sample?
5 Assume your class has been given the task of evaluating the methodology used in a case-control study to help to determine the cause of lung lead poisoning. Outline the advice you would provide to a fellow student who is unsure of how to carry out this task.
6 A scientist carried out a cohort study of the association between taking oral contraceptives and the risk of developing thrombosis. The steps followed in the study are outlined below.
   1 A group of women from the building in which the scientist worked took part in the study – approximately 350 women in total. None of these women suffered from thrombosis at the start of the study. Some took the contraceptive pill for the period of the study and others didn't.
   2 The scientist realised that she had more women who didn't take the oral contraceptive pill than those who did, so she removed enough of them to make equal numbers.
   3 At the initial meeting, the scientist gave each woman a scientifically valid questionnaire about many factors in her life, including whether she had thrombosis or not.
   4 Subsequent visits were carried out at yearly intervals, where valid questionnaires were again used and medical check-ups given, to determine whether thrombosis was present or not.
   5 This process was continued for five years and the results were statistically analysed using approved scientific and mathematical models.
   6 The study's results were then posted by the scientist on her colleague's website page.

   Evaluate the method used in this epidemiological study and suggest improvements that could be made to this study.

16.4 Benefits of epidemiological studies

The benefits of epidemiological studies are numerous and were outlined earlier in this chapter (page 542). Some of the types of epidemiological studies used, along with some examples of each, were provided on pages 554–6.

INVESTIGATION 16.5

A secondary-source investigation to evaluate the benefits of an epidemiological study

INTRODUCTION

A **benefit** is anything that provides an advantage to someone. Benefits can be short-term (such as being paid to walk the neighbour's dog), meaning that the advantage is immediate, or long-term (such as insurance), meaning that you might not receive the advantage for many years.

Benefits can also be direct or indirect. Direct benefits can be quantified, such as receiving pocket money for helping around the house. Indirect benefits are those that cannot be directly quantified but have an impact on you, such as your sister preparing your school lunch for you.

AIM

To evaluate the benefits of engaging in an epidemiological study

METHOD

Re-read the information about the Gila Indian River community (pages 558–60). Work in a group of 2–3 to brainstorm all the benefits that would have resulted from such a study. List these benefits in a results table like the one shown in the Results section. Within your group, decide whether the benefit is short- or long-term and direct or indirect.

RESULTS

Record your results in a table like the one below.

BENEFIT IDENTIFIED	TYPE OF BENEFIT (TICK APPROPRIATE BOX/BOXES)				WHO GETS THE BENEFIT?
	SHORT-TERM	LONG-TERM	DIRECT	INDIRECT	

DISCUSSION

1. Discuss the benefits of working as a team.
2. With reference to all the benefits you have identified, and the people who have been advantaged by these benefits, do you think that the Gila Indian River study provided value to those involved? Back up your claim with examples from the study.

CONCLUSION

Write a few summary sentences that relate to the aim of this investigation.

CHECK YOUR UNDERSTANDING 16.4

1. **a** List three major benefits of undertaking an epidemiological study.
   b State who gains the benefit in each case.
2. Distinguish between:
   a short- and long-term benefits
   b direct and indirect benefits.

16 CHAPTER SUMMARY

Epidemiology: Why are epidemiological studies used?

Epidemiology is the study of patterns of disease in populations.

Epidemiological study

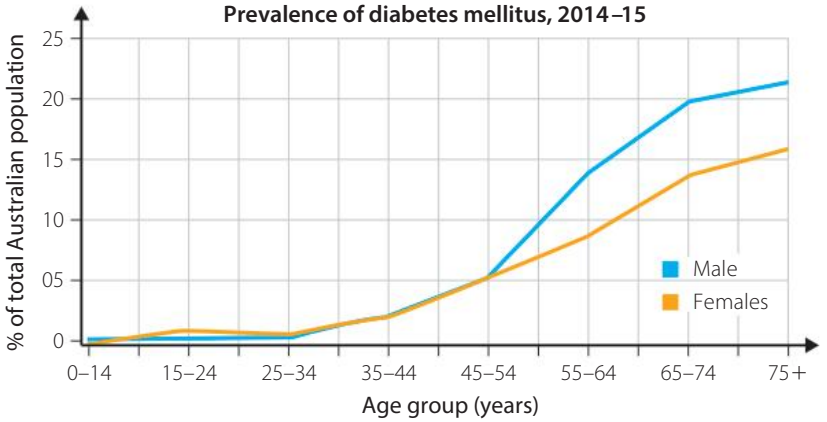
- Determines the cause of disease and which populations are affected
- Guides the development of strategies to control disease and improve public health
- Evaluates the effectiveness of strategies in place to treat/control disease

Analysis of data allows the identification of patterns and trends in the incidence, prevalence and mortality rates of disease.

Analysis of data to determine the populations affected

These graphs indicate that even though the incidence rate of colorectal cancer is only slightly higher for non-Indigenous populations than Indigenous populations, the mortality rate is much higher for non-Indigenous populations.

The prevalence of diabetes mellitus is 6% higher for males in the 65–74 age group than it is for females in the same age group.



Develop strategies to control disease

Melanoma treatment

Surgery
- Remove tumour

Radiation
- X-rays damage DNA of cancer cells

Chemotherapy
- Slows growth of cells
- Not as effective

Targeted therapies
- Drugs interrupt the pathways that cause uncontrolled cell division

Immunotherapy
- Checkpoint inhibitors cause the immune system to recognise and destroy melanoma cells

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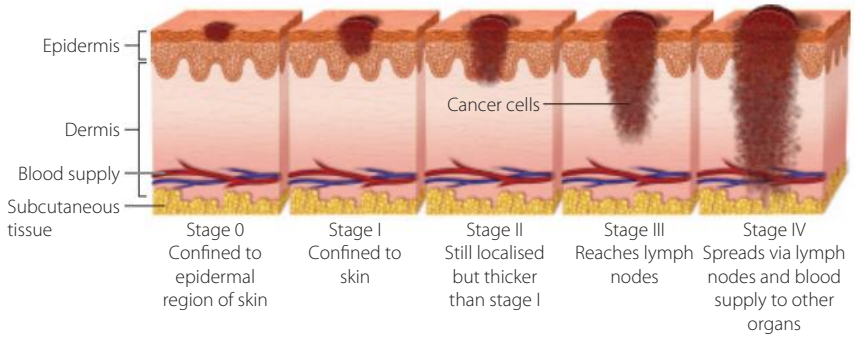


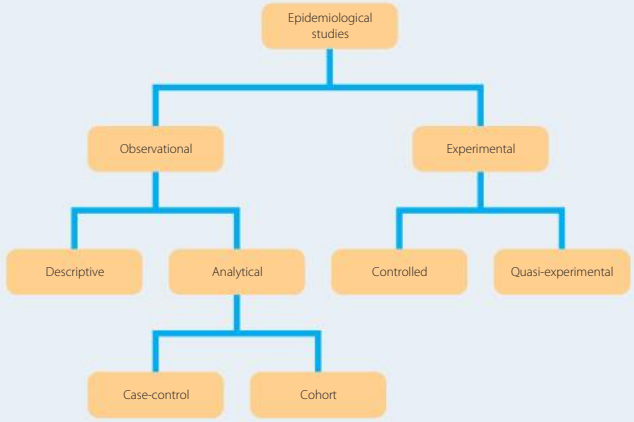
Future directions for melanoma treatment

Further research into targeted therapies, immunotherapies and vaccines.

Evaluate effectiveness of strategies to control disease

- Make a judgement based on criteria using evidence to support the judgement.
- The method used in study should follow accepted epidemiological principles. Errors should be included in evaluation.
- Criteria common to most epidemiological study include: large sample size, long period of study, using scientifically approved methods of implementing the study, collecting data and analysing results.

METHODS USED IN EPIDEMIOLOGICAL STUDIES



Descriptive studies

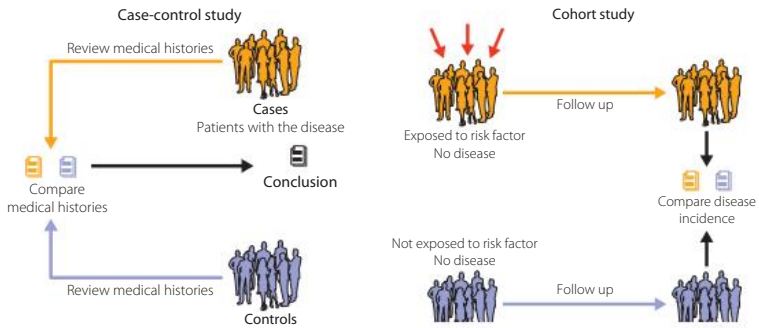
Collect information about frequency of the disease, population affected, location and time, and hypotheses are proposed from this

Analytical studies

Collect and analyse data

TYPES OF ANALYTICAL STUDIES

- **Case-control studies** – compare people who have the disease (cases) with people who do not have the disease (controls).
- **Cohort studies** – study two or more similar groups who are free of the disease. One group is exposed to the possible cause of the disease while the other group is not exposed.



Errors in epidemiological studies

Random errors

Unpredictable; reduce precision but do not skew the results

Systematic errors

Also known as bias – selection bias, information bias and confounding factors; shift the results of a study away from the true results

Intervention studies

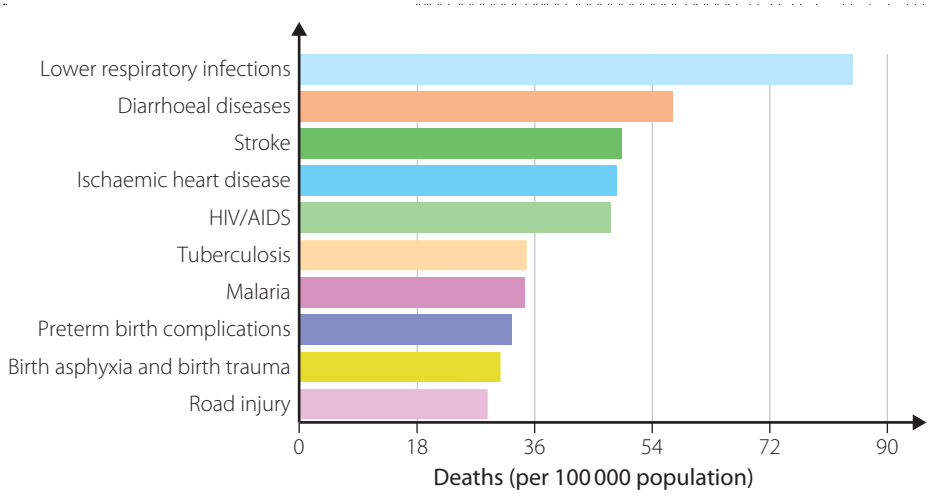
Test the effectiveness of a treatment for a disease, or the effectiveness of a public health campaign.

16 CHAPTER REVIEW QUESTIONS

Review quiz

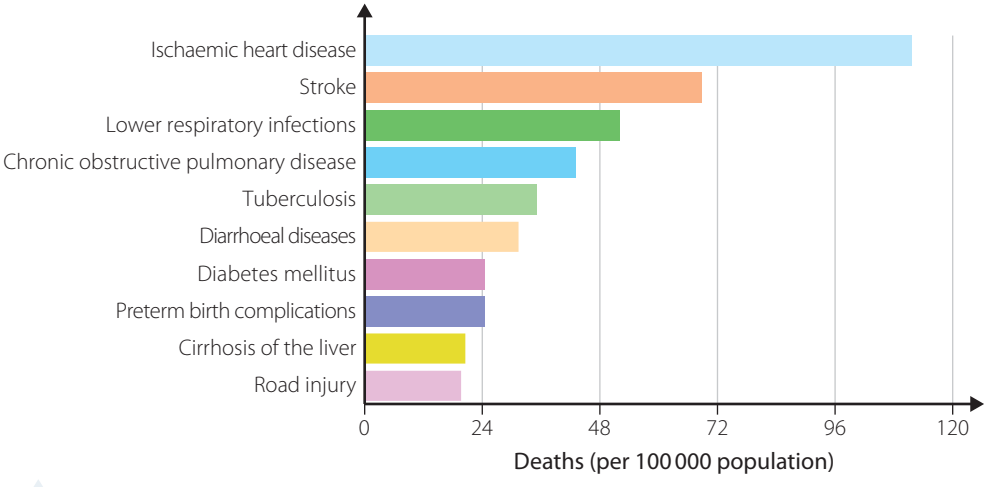
1 Experimental studies are often used to test the effectiveness of a new drug in the treatment of a disease.
   a Outline the recommended steps in the process when a study of this type is used for this purpose.
   b Discuss the ethical issues that may arise in relation to this type of study.

Questions 2–6 refer to the following figures.



Source: Global Health Estimates 2016. The Top 10 Causes of Death - Reprinted from Fact sheet N°310 – Updated May 2014. World Health Organisation, 2018 http://www.who.int/mediacentre/factsheets/fs310/en/

FIGURE 16.16 The top 10 causes of death in low-income economies, 2015



Source: Global Health Estimates 2016. The Top 10 Causes of Death - Reprinted from Fact sheet N°310 – Updated May 2014. World Health Organisation, 2018 http://www.who.int/mediacentre/factsheets/fs310/en/

FIGURE 16.17 The top 10 causes of death in low-middle income economies, 2015

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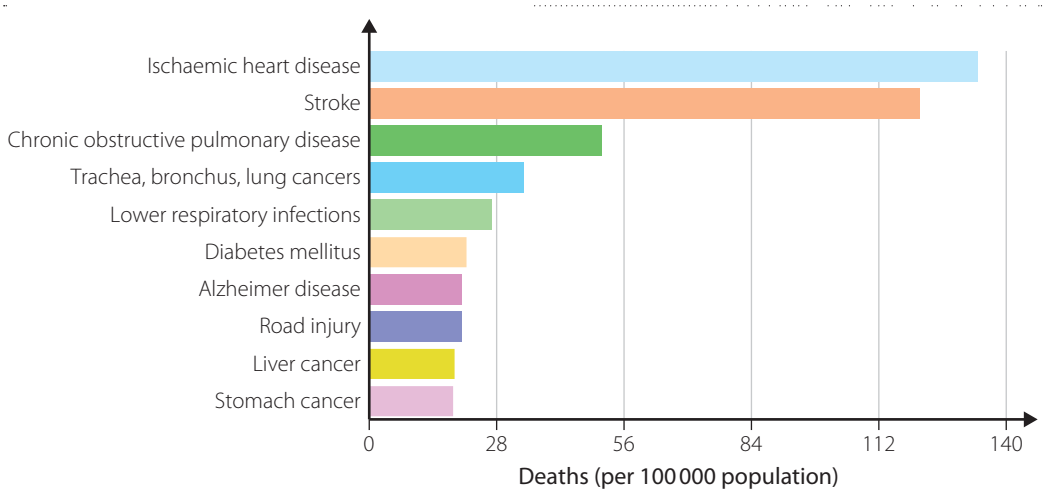
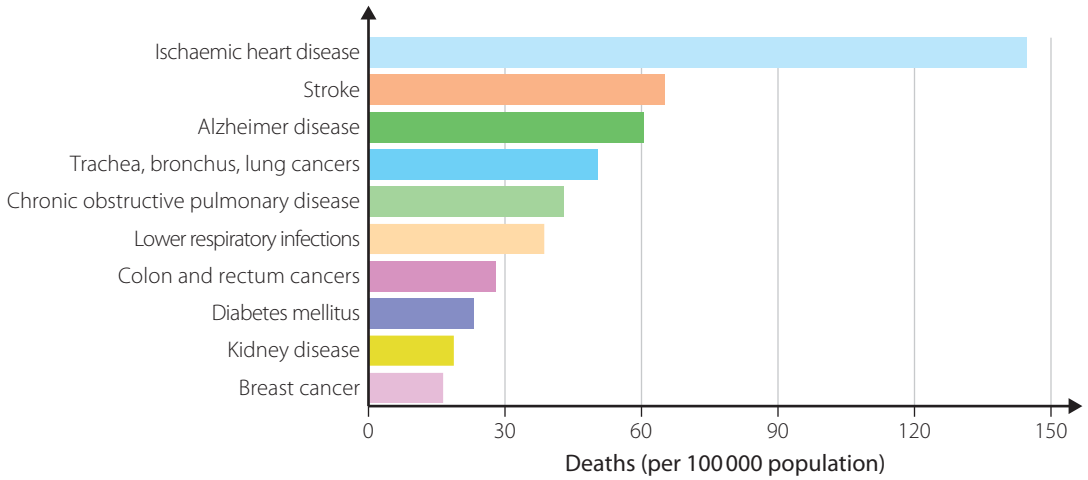


Global Health Estimates 2016: The Top 10 Causes of Death - Reprinted from Fact sheet N°310 – Updated May 2014. World Health Organisation, 2018 http://www.who.int/mediacentre/factsheets/fs310/en/

FIGURE 16.18 The top 10 causes of death in upper-middle-income economies, 2015



Source: Global Health Estimates 2016: The Top 10 Causes of Death - Reprinted from Fact sheet N°310 – Updated May 2014. World Health Organisation, 2018 http://www.who.int/mediacentre/factsheets/fs310/en/

FIGURE 16.19 The top 10 causes of death in high-income economies, 2015

2 Identify how many infectious diseases and how many non-infectious diseases are in the top 10 causes of death for each income group. Present your answer in a table.

3 Compare the top 10 causes of death in each income-economy group.

4 Discuss possible reasons for the similarities/differences observed in Question 3.

5 How could you check the accuracy of your proposed reasons in Question 4?

6 Explain why it is important to collect data for different groups of people.

7 Table 16.2 shows the prevalence of obesity as a percentage of the total population for age and gender in Australia in 2014–2015. The population aged over 18 in Australia at this time was approximately 17.9 million.

TABLE 16.2 Percentage obesity by age and gender, Australia, 2014–2015

AGE GROUP (YEARS)	PROPORTION OF INDIVIDUALS WHO ARE OBESE (%)		
	MALES	FEMALES	TOTAL
18–24	17.3	17.3	17.1
25–34	20.8	17.3	19.0
35–44	26.7	30.7	28.6
45–54	33.2	33.0	33.0
55–64	36.8	34.9	35.9
65–74	38.2	32.7	35.4
75–84	32.4	27.1	29.6
85+	11.2	21.5	17.8
Total	**28.4**	**27.4**	**27.9**

Source: Australian Bureau of Statistics (ABS). Australian Health Survey 2014/15 (4364.0) CC BY-2.5 Australia licence

a Using the value given for the total population over 18 in 2014–2015, calculate:
  i the number of males and females over 18 who were obese
  ii the total number of Australians over 18 who were obese.
b Identify the age group with the highest proportion of obese individuals, for males, females and total.
c Draw a line graph from the information presented in Table 16.2. Use a key to indicate which line represents males, females and the total percentage.
d From this line graph, outline the trends by age group shown in the prevalence of obesity for males and females.
e Describe how this information could be used to improve the health of the population.

8 The incidence of a type of non-infectious disease (called P disease) has increased in particular areas over the last few years. Initial descriptive studies have been carried out and it has been hypothesised from these results that the cause of P disease is exposure to pollutants in a body of water that supplies a limited number of houses and factories in a rural city. Design a case-control study that could be used to test this hypothesis.

9 Prepare an answer to the Inquiry question: Why are epidemiological studies used?

Exam preparation

17 Prevention

INQUIRY QUESTION

How can non-infectious diseases be prevented?

Students:

- use secondary sources to evaluate the effectiveness of current disease-prevention methods and develop strategies for the prevention of a non-infectious disease, including but not limited to: CCT
  - educational programs and campaigns PS
  - genetic engineering EU

Chinnapong/Shutterstock.com

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Assessments
- Chapter review
- Review quiz
- Exam preparation

Investigations
- **17.1** A secondary-source investigation and evaluation of an educational program and public health campaign
- **17.2** A secondary-source investigation into prevention of non-infectious disease using genetic engineering
- **17.3** A secondary-source and first-hand investigation to develop strategies to prevent a non-infectious disease outbreak

Worksheets
- Prevention of a genetic disease by pre-implantation genetic testing
- Prevention of a nutrition disease by genetic engineering: golden rice
- Prevention of an environmental disease by public health campaigns: skin cancer
- Prevention of non-infectious diseases

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

Source: Foundation for Alcohol Research and Education

FIGURE 17.1 Prevention of disease is always preferable

Prevention of disease is always a much better option than suffering the debilitating effects of a disease or its treatment. Treatments often involve the use of drugs, many of which have side effects that cause more problems. Some diseases, such as cystic fibrosis, have no effective treatment, meaning that the only option is to manage and reduce the severity of the symptoms. With these diseases, the quality of life of affected individuals is compromised and there is a continuing financial burden on the individual, their family and the health system.

Strategies to prevent non-infectious diseases and disorders are important, not only to reduce the suffering of individuals and improve their quality of life, but also to improve the overall health and wellbeing of the population. Prevention also reduces the financial burden on both the individual and the health system, allowing resources to be directed to other areas (Fig. 17.1).

17.1 Educational programs and campaigns

Many preventable non-infectious diseases are caused by nutritional imbalances or exposure to environmental factors, including lifestyle factors (see Chapter 15). Data obtained from epidemiological studies identifies the diseases that are most prevalent in populations and the groups that are most at risk. This assists health authorities and governments to develop strategies aimed at preventing the diseases that are of most concern because of their incidence, prevalence and mortality.

Strategies for prevention are varied and include educational programs and public health campaigns, genetic engineering and government legislation. These include financial levies imposed on substances that are thought to be detrimental to health.

Educational programs and campaigns use strategies to provide information and educate the population about the effects of a disease and the risk factors that increase the chance of developing that disease. Suggestions about how to avoid the risk factors are a part of these programs and campaigns, educating members of the population in the hope that they will change their behaviour to reduce their exposure to these risk factors and lower their chances of developing the disease (Fig. 17.2).

A successful public health campaign has a number of key components (Fig. 17.3):

- an evidence base for action
- a package of a limited number of high priority evidence-based interventions
- effective performance management with real-time monitoring, evaluation and program improvement
- partnerships between the public and private sectors
- communication of accurate information to healthcare workers and the public
- political commitment.

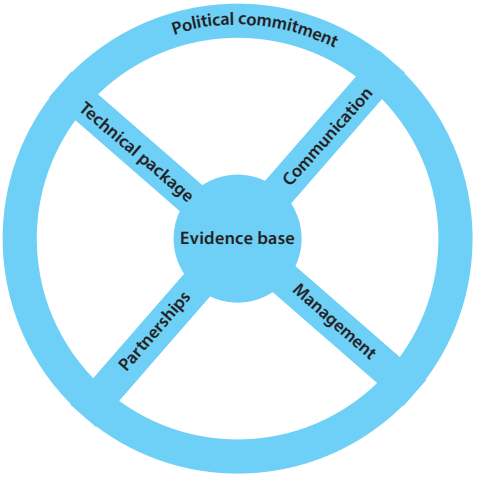
There are many examples of educational programs and public health campaigns that have been implemented in Australia to change the behaviour of members of the population (Fig. 17.4). These include the QUIT program, the National Alcohol Strategy, the Diabetes Helpline, 'Slip, Slop, Slap, Seek, Slide', 'Kids Matter', 'Jump Rope For Heart', 'Alcohol. Think again', the Needle and Syringe Program, and the Pregnancy, Birth and Baby helpline. These campaigns were developed as a result of national strategic plans aimed at preventing common and emerging diseases.

FIGURE 17.2 Prevention of non-infectious diseases is closely linked to following a healthy lifestyle.

Legislation

Non-infectious diseases are a major cause of death, disease and disability in the population. This places a strain on the health system and economic development, and affects the wellbeing of many individuals in a population.

The main risk factors associated with non-infectious disease are tobacco, alcohol, unhealthy diet and lack of physical activity. A major challenge for people who have the desire to change their behaviour is the addictive nature of some of these risk factors. Educational programs and public health campaigns are often not enough to change the behaviour of members of the population. Therefore it is essential that governments introduce **legislation** to minimise these risk factors in the population.



Source: National Center for Biotechnology Information, U.S. National Library of Medicine

FIGURE 17.3 The six key criteria for an effective public health campaign

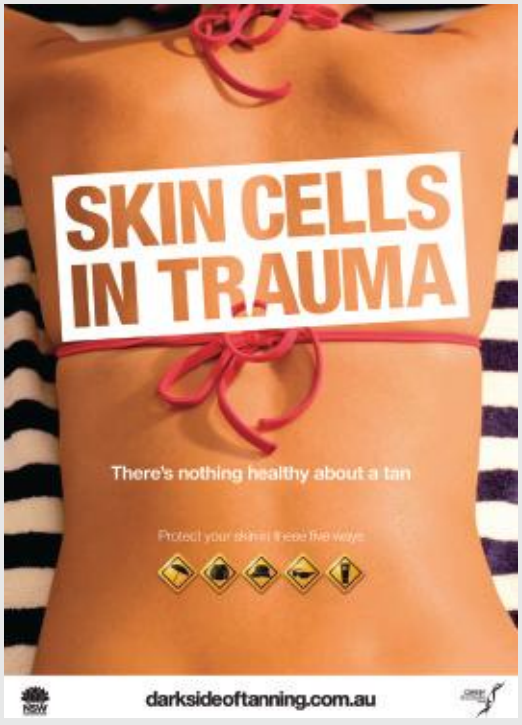
This legislation could be in a number of forms, to target different aspects of the specific risk factor. It could prohibit the promotion and marketing of the use of the risk factor, including a ban on advertising and sponsorship. It could increase tax (**levy**) on the risk factor, to deter the purchase of products related to the risk factor. For example, legislation to tax alcohol and tobacco products is in place to deter the purchase of these products. There is currently a push for a 'sugar' tax to be introduced, to deter people from consuming too many sugary soft drinks (Fig. 17.5, page 574). Legislation is also used to restrict the places and times in which the risk factor can be used – for example, a ban on alcohol consumption in certain areas and restrictions on the operating hours of bars, bottle shops and hotels. Legislation has also been introduced to enforce the clear labelling of food products in supermarkets and the kilojoule content of take-away foods.

Some of the legislation that has been introduced by the government to reduce the use and effects of tobacco include:

- a ban on all tobacco advertising, promotion and sponsorship
- a ban on smoking in the workplace and in public places
- plain paper packaging of cigarettes, containing pictorial, graphic warnings and no logos, colours, brand images or promotional information
- an increase in taxes on tobacco products.

Components of public health campaigns

- National Days or weeks to raise awareness and educate people about a disease, such as National Diabetes Week, Skin Cancer Action Week, Heart Week
- Online resources to assist individuals and groups in implementing lifestyle behaviour changes
- Advertising campaigns that saturate all forms of media, including social media, detailing effects of a disease and risk factors that increase the likelihood of developing the disease. These include catchy slogans and sometimes graphic images of the effects of the disease on the body, e.g. the risk of exposure.
- The use of social media and influencers
- Apps that can be downloaded to assist with reducing exposure to risk factors
- Posters and advertising boards in many public places, including roadside (e.g. bus stops), sporting stadiums, railway stations or other places where large numbers of people will see them.
- Tailoring the delivery of the message to the target audience for maximum engagement , e.g. involving sporting teams in campaigns, such as 'Quit B Fit' slogan on football players' shorts
- Screening those at risk for factors that indicate an increased likelihood of developing the disease, e.g. screening for pre-diabetes
- National helplines, such as the Quitline
- Support programs such as dietary and physical activity advice for those who are overweight and obese
- Legislation to reduce risk factors associated with non-infectious diseases/disorders, e.g. lockout laws to reduce the risk of alcohol-related injuries leading to brain injuries, plain paper packaging on cigarettes and restriction of all cigarette advertising in all forms including online.
- Funding for organisations that provide support services to educate and support those trying to change their lifestyle behaviours, including the Heart Foundation, Diabetes Australia, Cancer Council Australia and many more
- Excise on some products, such as alcohol and cigarettes, as a deterrent to buying these products



© Cancer Institute NSW

Cancer Council Victoria

Source: Diabetes Australia

FIGURE 17.4 Examples of health campaigns in Australia

Source: Lung Foundation Australia

Jeffrey Blackler / Alamy Stock Photo

© Commonwealth of Australia, Department of Health, National Tobacco Campaign

Foundation for Alcohol Research and Education

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Impact of alcohol

Discuss how this animation may help motivate someone to reduce their alcohol intake.

FIGURE 17.4 (*Continued*)

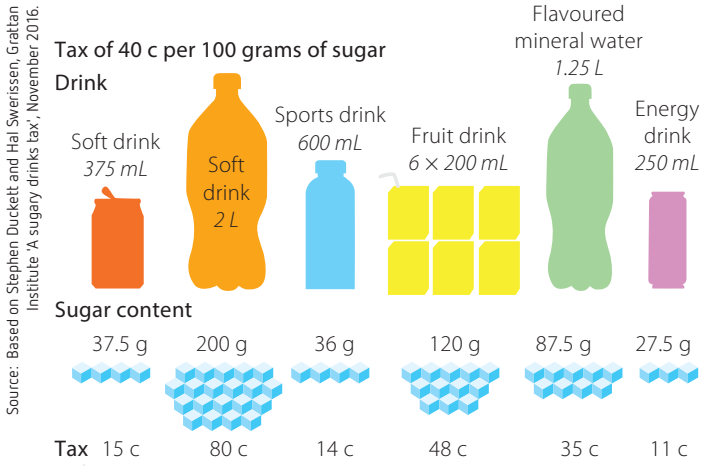


FIGURE 17.5 The impact of the proposed sugar content tax on retail prices of sugary drinks

KEY CONCEPTS

- Prevention of disease is always the preferred option.
- Prevention reduces the burden of disease – physical, financial and psychological – on the individual and the health system.
- Changing the behaviour of individuals will prevent many non-infectious diseases.
- Strategies to change behaviours include education programs and campaigns, legislation, support groups, and targeting those at risk.
- Epidemiological studies provide data to identify areas of risk, to direct strategies for prevention.

CHECK YOUR UNDERSTANDING 17.1a

1. Outline the advantages of preventing non-infectious disease.
2. How does changing the lifestyle behaviour of individuals help to prevent non-infectious disease?
3. Identify some strategies in common use to prevent non-infectious disease.
4. What are three pros and three cons of the introduction of a sugar tax to deter people from drinking sugar-laden soft drinks?
5. Describe the role of epidemiological studies in assisting the prevention of non-infectious disease.

Evaluating the effectiveness of educational programs and campaigns

Comparing incidence and prevalence data before and after the implementation of educational programs and public health campaigns can provide a measure of the effectiveness of the strategies employed.

QUIT campaign

Weblink
Cancer Council
Watch the video and follow the interactive that educates people about cancer and its prevention. Outline how these would help to prevent cancer.

Smoking remains the leading preventable cause of death in the Australian population, with 15 500 smoking-related deaths each year. The QUIT campaign was developed as an educational program and campaign to reduce the prevalence of smoking-related diseases in the population.

Lung cancer is just one of many smoking-related diseases, which include many types of cancers, heart disease and other lung diseases. Many epidemiological studies have been carried out to determine the cause of lung cancer, and the findings have universally demonstrated a clear link between smoking and the increased incidence of lung cancer (Fig. 17.6). The studies have also shown a clear link between cigarette smoking and reduced life expectancy.

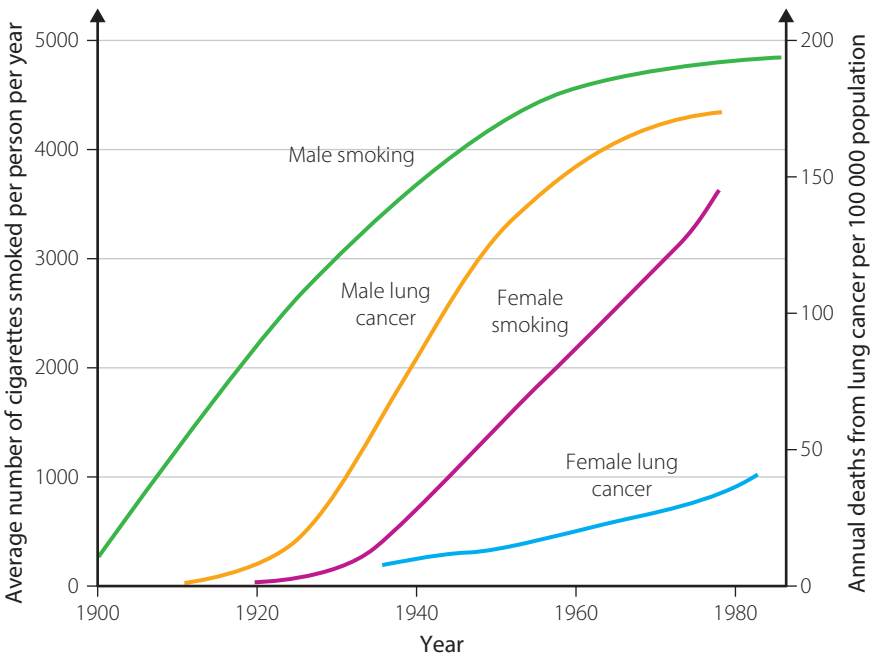


FIGURE 17.6 Data from long-term epidemiological studies shows a direct link between smoking and lung cancer.

Studies comparing smokers and non-smokers show that smokers have a 10 times greater chance than non-smokers of dying from lung cancer. Further studies show that the more cigarettes smoked each day, the greater the incidence of lung cancer. It has also been shown that the longer a person smokes, the greater their chance of developing lung cancer. Exposure to passive smoking also increases the risk of developing lung cancer.

The campaign to reduce smoking and, hence, smoking-related diseases has a multifaceted approach that involves education to raise awareness of risk factors, as well as legislative changes. The QUIT program has evolved over many years, and runs in conjunction with many strategies, including:

- the use of slogans such as 'Quit for life', 'Quit B fit' and 'iCanQuit' (Fig. 17.7a)
- graphic images in the media and on cigarette packets to highlight the dangers of cigarette smoking (Fig. 17.7b)
- showing people with lung cancer in their own life situations, expressing the wish that they had given up smoking earlier so that their life was not cut short
- a national helpline to support smokers in their quest to change their behaviour and break the smoking habit

a

Wellington Aboriginal Corporation Health Services National Tackling Indigenous Smoking Program

b

AP/Minister for Health and Ageing/AAPImage

FIGURE 17.7 Strategies used in the QUIT campaign: **a** Quit B Fit campaign, part of the Indigenous anti-smoking program; **b** plain paper packaging with graphic images of the effects of smoking

- legislation requiring plain paper packaging on all cigarette packets and storage of cigarette packets in cupboards out of sight of consumers, to reduce the appeal of tobacco products to consumers and prevent tobacco companies using the packaging to downplay the harmful effects of tobacco
- increased excise on tobacco products to make them more expensive, in a bid to discourage consumers from buying them
- legislation to restrict advertising, including Internet advertising, of tobacco products
- raising the legal age for smoking to 21 (currently under consideration by the Australian Government).

The campaign to reduce the smoking rate in Australia and, hence, reduce the prevalence, incidence rate and mortality rate of smoking-related diseases such as lung cancer is a long-term one. The list below outlines the strategies put in place by governments and public health organisations over the past 44 years.

- 1973 – health warnings first mandated on all cigarette packs in Australia
- 1976 – bans on all cigarette advertising on radio and television in Australia
- 1986 to 2006 – phased in bans on smoking in workplaces and public places
- 1990 – bans on advertising of tobacco products in newspapers and magazines published in Australia
- 1992 – increase in the tobacco excise
- 1993 – *Tobacco Advertising Prohibition Act 1992* prohibited broadcasting and publication of tobacco advertisements
- 1994 to 2003 – bans on smoking in restaurants
- 1995 – nationally consistent text-only health warnings required
- 1998 to 2006 – bans on point-of-sale tobacco advertising across Australia
- 2006 – graphic health warnings required on packaging of most tobacco products
- 2010 – 25% increase in the tobacco excise
- 2011 – first complete state or territory ban on point-of-sale tobacco product displays
- 2012 – offence for any person to publish tobacco advertising on the internet or other electronic media
- 2012 – introduction of tobacco plain packaging, and updated and expanded graphic health warnings
- 2012 – reduction in the duty free allowance from 250 cigarettes or 250 grams of cigars or tobacco products to 50 cigarettes or 50 grams of cigars or tobacco products from 1 September 2012
- 2013 – first 12.5% tobacco excise increase on 1 December.
- 2014 – change from bi-annual indexation based on the Consumer Price Index (CPI) to bi-annual indexation based on average weekly ordinary time earnings (AWOTE)
- 2014 – 12.5% excise increase on 1 September
- 2015 – 12.5% excise increase on 1 September
- 2016 – release of the Post Implementation Review of Tobacco Plain Packaging
- 2016 – 12.5% excise increase on 1 September
- 2017 – additional four annual 12.5% tobacco excise increases implemented on 1 September each year from 2017 to 2020 inclusive
- 2017 – reduction in duty free tobacco allowance, 25 grams of duty free tobacco (cigarette, loose leaf etc), plus one open packet; equivalent to approximately 25 cigarettes
- 2017 – harmonisation of the taxation of roll-your-own tobacco and other products such as cigars, with manufactured cigarettes

(Source: Australian Government, Dept of Health 2017, 'Tobacco control timeline', http://www.health.gov.au/internet/publications/publishing.nsf/Content/tobacco-control-toc~timeline)

The graph in Figure 17.8 shows changes in the prevalence of smoking in the Australian population from 1990 to 2013, in relation to the strategies implemented to control tobacco products since 1990.

This graph indicates that strategies implemented to control the prevalence of smoking in the Australian population have been somewhat successful, as the rate of smoking reduced from just over 24% in 1991 to approximately 13% in 2013. It is predicted that the prevalence of smoking will decrease further with the continuation and expansion of these strategies.

As well as statistics showing the decrease in smoking prevalence due to the QUIT campaign and other government strategies, statistics on the incidence of disease caused by smoking can be studied to further *evaluate* the effectiveness of these strategies. The incidence and mortality rates from lung cancer are shown in Figure 17.9.

Evaluate means to judge the quality, importance or value of something.

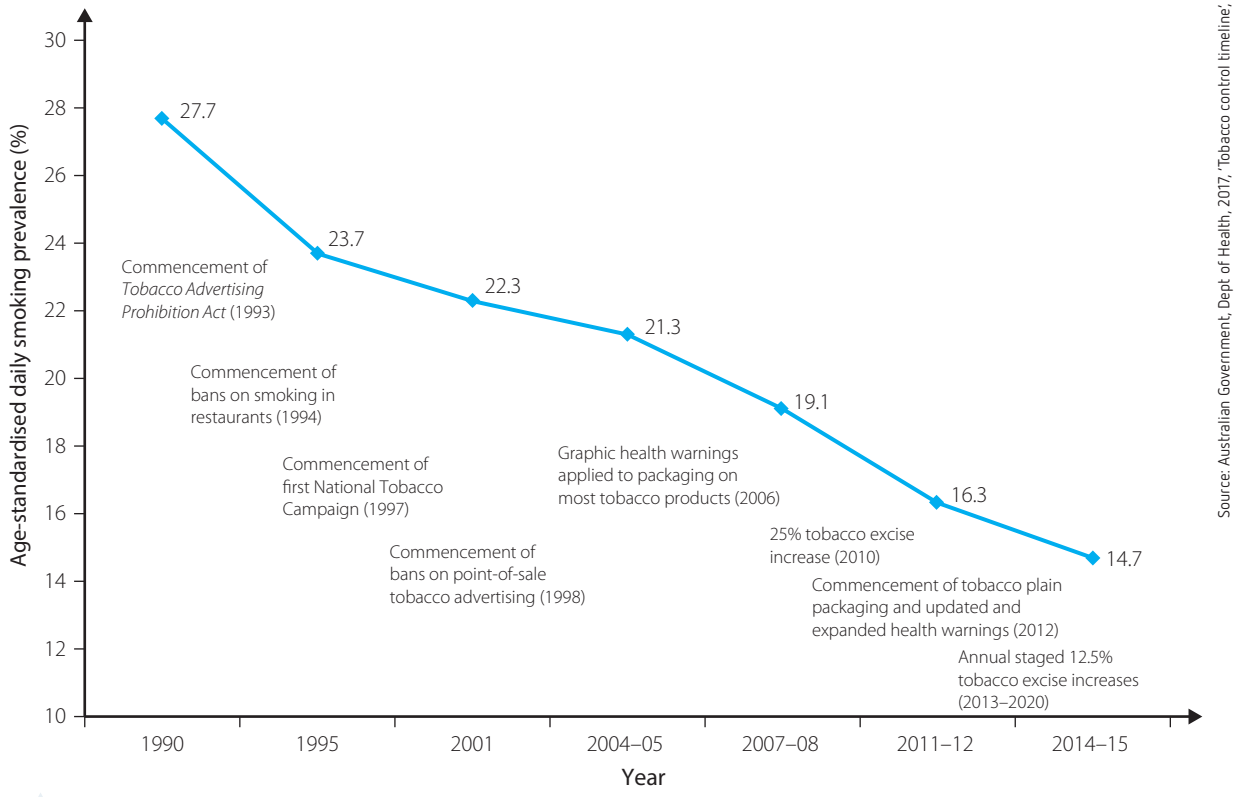


FIGURE 17.8 Age-standardised daily smoking prevalence in the general population of people aged 18 years and older and key tobacco control measures implemented in Australia since 1990

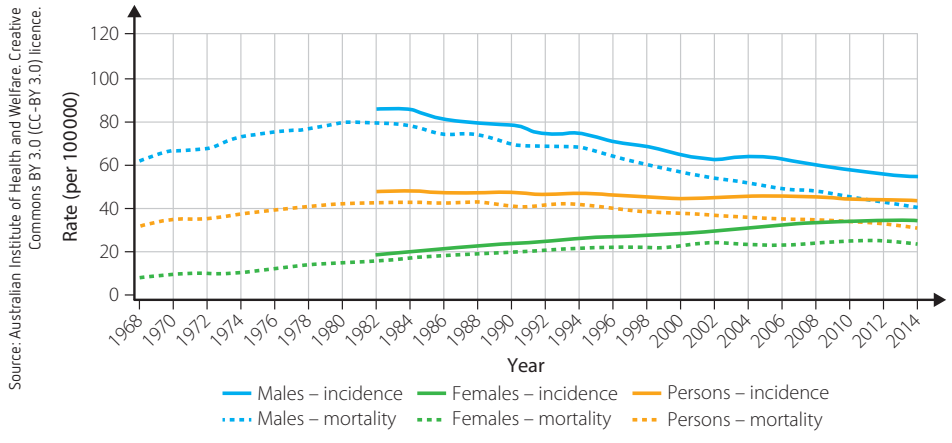


FIGURE 17.9 Age-standardised incidence rates for lung cancer 1982–2013 and age-standardised mortality rates for lung cancer 1968–2014, by sex

The graph shows an initial increase in the mortality rate from lung cancer for males from 1968 to 1982, followed by a decrease in the mortality rate from 1982 to 2014. It also shows a gradual increase in the mortality rate from lung cancer for females from 1968 until the rate becomes relatively steady from 2002.

Figure 17.9 shows that males have a higher mortality and incidence rate for lung cancer than females. It also shows that, for males, the incidence rate per 100 000 for lung cancer has decreased steadily since 1982. It is predicted that the rate will continue to decrease.

The female incidence rate of lung cancer per 100 000 was much lower than the rate for males in 1968, but showed a steady increase until 2014, when it was much closer to males. Again, compared to males, the female mortality rate was much lower in 1968, but increased steadily until 2014, when the difference in mortality rates between males and females was much closer.

The difference between the trends for males and females could be explained by the fact that lung cancer takes several decades to develop (as shown in Fig. 17.6). While the rate of smoking is decreasing now for both males and females, the decrease in the rate of smoking occurred earlier in males (1960s) than in females (late 1970s).

The rate of smoking is predicted to continue to decrease for both sexes. While the incidence and mortality rates from lung cancer are also predicted to continue to decrease for males, the incidence and mortality rates for females are predicted to increase before eventually decreasing.

These statistics show that the education programs, public health campaigns and legislation have been successful in reducing the rate of smoking in both males and females. As a result of this decrease in smoking rates, the incidence and mortality rates of lung cancer in males have decreased, and are predicted to also decrease for females.

While the rate of smoking has decreased, there has been an increase in the use of e-cigarettes, by young people especially. The rationale behind the use of e-cigarettes is that people think that they are safer to use than tobacco products and/or that their use may help them to quit smoking.

Electronic cigarettes

An **electronic cigarette** (e-cigarette) is a battery-powered device that changes a liquid (**e-liquid**) into an aerosol that is then inhaled into the person's lungs (Fig. 17.10). With a conventional cigarette, smoke from burning tobacco is inhaled into the lungs, whereas the aerosol inhaled from an e-cigarette is a vapour in which liquefied nicotine or other chemicals are dissolved.

It is illegal to sell e-cigarettes and accessories to minors under the age of 18, to buy e-cigarettes and

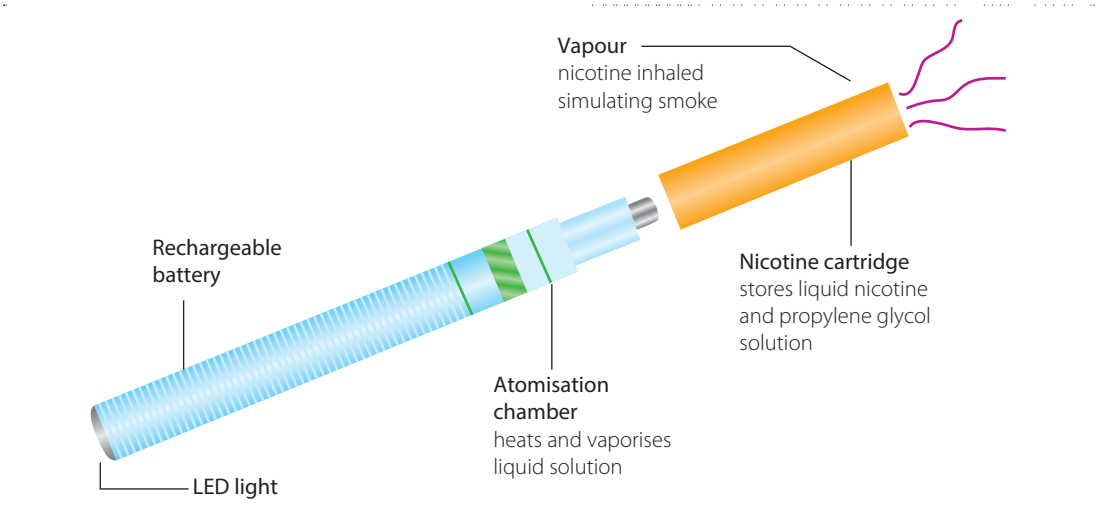


FIGURE 17.10 The components of an e-cigarette

accessories for minors under the age of 18, or to smoke e-cigarettes in cars with children under the age of 16 present. It is also illegal to sell liquid nicotine, including e-liquids containing nicotine.

How safe it is to use e-cigarettes is unknown at present, as there is little information available. Many e-liquids that claim to be free of nicotine have been found on analysis to contain varying amounts of nicotine. Nicotine has many short- and long-term deleterious effects on the body, and if liquid nicotine is ingested it can cause poisoning. The effects on the body of the other chemicals in the e-liquid are as yet unclear.

Some manufacturers of e-cigarettes claim that they can help a person quit smoking, but this has not been demonstrated scientifically. Health authorities have not approved e-cigarettes as an aid to quitting smoking or as a nicotine replacement therapy.

INVESTIGATION 17.1

A secondary-source investigation and evaluation of an educational program and public health campaign

INTRODUCTION

In this investigation you will research information and evaluate a current educational program/campaign designed to help prevent a non-infectious disease/disorder.

AIM

To investigate and evaluate a current non-infectious disease prevention method involving an educational program and public health campaign

METHOD

1. Working collaboratively, research a current educational program/campaign aimed at preventing a non-infectious disease.
2. Address the following areas:
   - Identify the program/campaign.
   - Identify the disease/disorder that it is designed to prevent.
   - Outline the strategies that are used to try to change the behaviour of individuals in the population.
   - Include any other relevant information about the program/campaign.
3. Statistics on the incidence, prevalence and mortality rate of the disease/disorder you are investigating. These statistics should be sourced for time periods both prior to and after the introduction of the program/campaign.
4. Use this and any other relevant information to evaluate the effectiveness of your chosen program/campaign in preventing the non-infectious disease/disorder it targeted.
5. Refer to page 571 to see the six key criteria for a successful public health campaign. How does your chosen campaign measure up against these criteria?
6. Present your information in a manner that makes the data and your evaluation clear and easy to understand.

RESULTS

Address all areas indicated in the Method steps above, in a manner that you think most clearly conveys information about the disease/disorder, the program that has been implemented to prevent it, and your evaluation of the effectiveness of the program in preventing this disease.

In this investigation, revise and use the same evaluation techniques you used in the previous chapter.

Critical and creative thinking

Literacy

Information and communication technology capability

Personal and social capability

Use the QUIT campaign (discussed in the previous section) for examples of the types of data/statistics that can be sourced.

»

» DISCUSSION

1 Outline why educational programs/campaigns are necessary.
2 Discuss how data can be used to evaluate the effectiveness of a campaign.

CONCLUSION

Write a few summary sentences to address the aim of this investigation.

KEY CONCEPTS

- Data from epidemiological studies can be used to evaluate the effectiveness of strategies such as education programs and public health campaigns.
- Comparison of data related to incidence, prevalence and mortality rates before and after the implementation of strategies can be used to evaluate their effectiveness.
- Implementation of strategies to reduce the prevalence of smoking in Australia have been effective in reducing the incidence and mortality rates of lung cancer.

CHECK YOUR UNDERSTANDING 17.1b

1 Define 'evaluate'.
2 Outline how data can be used to evaluate the effectiveness of strategies introduced to prevent non-infectious disease.
3 Describe the trend in the prevalence of smoking in Australia since 1990.
4 a Compare the trends in lung cancer mortality rates for males and females in Australia from 2006 to 2015.
  b Link these trends to the trends shown for smoking prevalence.
5 Discuss whether the strategies implemented to reduce the prevalence of smoking have been effective.
6 Analyse the QUIT campaign and categorise each strategy under the six key criteria for a successful public health campaign listed on page 571. Evaluate the campaign using these criteria.

17.2 Genetic engineering

Refer to the discussion of inheritance patterns in Chapter 6.

Genetic engineering was discussed in Chapters 8 and 9.

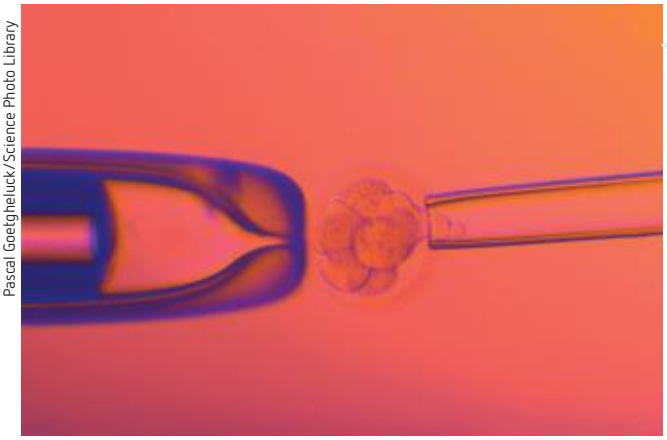
The ability to manipulate genes has enabled us to devise a number of ways to prevent certain non-infectious diseases/disorders. Genetic engineering techniques used in processes such as **pre-implantation genetic testing** (PGT) can prevent genetic disease, while the use of transgenic crops as food sources has the potential to prevent some types of nutritional disease. Genetic engineering techniques are used to produce vaccines such as Gardasil®, to prevent human papilloma virus (HPV) infection, which in turn prevents cervical cancer. Genetic engineering often brings with it debate on the ethics of its use (see Chapter 8), which needs to be balanced with its effectiveness in the prevention of disease.

Pre-implantation genetic testing

A young couple's first child was born without any problems but they soon learned that their baby son was profoundly deaf. With genetic testing it was discovered that their baby was homozygous recessive for a mutation of a gene that is involved in the proper functioning of the cochlea.

Genetic counselling followed, and the couple were informed that any future children had a 25% chance of being homozygous recessive for this mutated gene and therefore of being deaf.

For the next birth, the couple decided they would undergo pre-implantation genetic testing (PGT). This began with a cycle of IVF (in vitro fertilisation), where eggs were harvested and collected. The eggs were then fertilised by direct injection of the sperm, and the embryos were grown for three days. One cell from each embryo was then removed (Fig. 17.11) and tested for the mutated gene. Embryos that were free of the gene or were carriers were retained, while the other embryos were either destroyed or used for research.



Pascal Goetgheluck/Science Photo Library

FIGURE 17.11
Removal of a cell from an embryo for pre-implantation genetic testing

On day 5, one of the retained embryos was implanted, while the rest were frozen for later use. The pregnancy developed normally and the baby was born with normal hearing. Through the use of PGT, the chance of the couple's offspring being born deaf was eliminated.

Genetic diseases/disorders can be caused by hereditary factors. If parents are aware or become aware that one or both possess an allele for a certain genetic disease/disorder (as the couple above did), they may undergo genetic counselling to determine the percentage chance of any offspring inheriting the condition. The parents then have a number of options available to them.

The couple decided that the chances of a child being born deaf was too great when conceived naturally and didn't want to take that chance. They thought about not having any more children, to prevent any occurrence of this disorder, but they wanted to expand their family.

This couple, after much research and discussion, decided to have children using the reproductive technology technique known as IVF (in vitro fertilisation), combined with PGT. This process allows couples at risk to ensure that their children are born free of the disease without having to go through the process of testing the foetus and then having to make the difficult decision about whether to go ahead with the pregnancy or terminate it if the disease/disorder is present.

Pre-implantation testing involves the fertilisation of the mother's egg with the father's sperm in an external laboratory setting (IVF), the removal of a single cell from an eight-cell embryo, and the use a genetic engineering technique known as **array comparative genomic hybridisation** (aCGH) to test the cell for the presence of the known genetic condition (Fig. 17.12). This technique can also determine whether the embryo is a carrier of the condition or free of any copies of the mutated allele. Embryos can also be tested for chromosomal abnormalities. Embryos that are free of the genetic disease/condition or are carriers are retained for implantation. In this way, the genetic disease/disorder is prevented in any offspring of the couple.

A wide range of single-gene disorders can be tested for, including cystic fibrosis, Huntington disease, thalassaemia and muscular dystrophy. This method, which combines the use of a reproductive technology (IVF) and genetic engineering to test the cells of the embryo (PGT), is 100% effective in preventing the disease.

There are many ethical issues associated with the use of reproductive and genetic engineering technologies.

Weblink
Process of PGT
Watch the video to see an outline of the process of PGT and some of the issues associated with its use.

Worksheet
Prevention of a genetic disease by pre-implantation genetic testing

Ethical issues involved with the use of these technologies are addressed in Chapters 8 and 9.

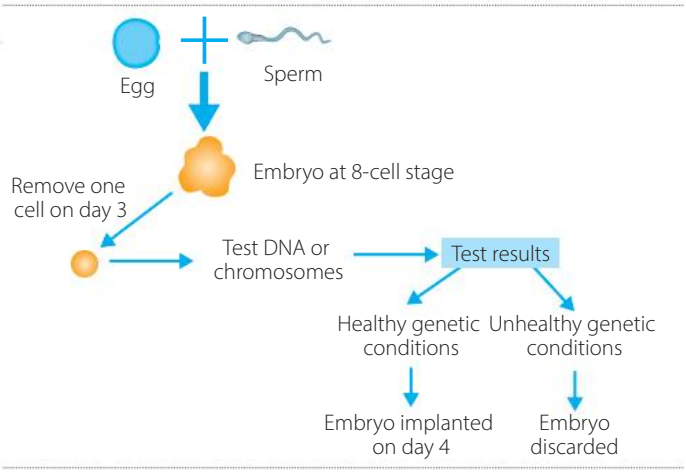


FIGURE 17.12 The process of pre-implantation genetic testing (PGT)

Transgenic species were discussed in Chapter 9, pp. 295–8.

Using genetic engineering to prevent nutritional diseases

Populations in developing countries often experience diseases/disorders caused by vitamin deficiencies, because of a poor diet. Researchers have been investigating the use of transgenic crops to incorporate vitamins into their staple foods. If successful, this would prevent the disorders/conditions caused by lack of a particular vitamin.

Refer to page 268 for more information on golden rice.

One example of this is the research carried out to produce what is known as 'golden rice'. This is rice that has been genetically engineered to include a gene from maize and a gene from a soil bacterium. This allows the rice to produce beta-carotene, which is then used by the body to produce vitamin A. The beta-carotene gives the rice its characteristic yellow colour (Fig. 17.13).

A deficiency of vitamin A in the diet can lead to blindness and a compromised immune system. In Africa and Asia it is estimated that approximately a quarter to half a million children suffer from blindness caused by vitamin A deficiency. In addition to this, two million people die each year of diseases they would otherwise have survived, because of their weakened immune system.

FIGURE 17.13 Golden rice is yellow and is here compared to normal rice. The yellow colour comes from the insertion of a gene to produce beta-carotene, which is used by the body to produce vitamin A.

Golden rice has been under development for many years and has suffered setbacks, due to a smaller yield being produced compared to normal rice and opposition to genetically modified foods from organisations such as Greenpeace. The Bill and Melinda Gates Foundation is supporting the final testing of golden rice and it was released in Bangladesh in 2018.

If successful, golden rice will prevent diseases and disorders caused by lack of vitamin A.

Weblink
Golden rice
Watch the video to understand the development of golden rice to counteract vitamin A deficiency, and the debate about its use.

Worksheet
Prevention of a nutritional disease by genetic engineering: golden rice

Refer to Chapter 5, page 185 for information about SNPs, haplotypes and GWAS.

Genetic engineering as a screening tool for disease susceptibility

Genetic engineering techniques are being used to generate information about non-infectious disease that is proving to be very valuable in promoting efficient disease prevention and management strategies. Both mapping of the human genome (through the Human Genome Project) and Genome Wide Association Studies (see Chapter 5) have contributed to a better understanding of the genetic basis of non-infectious disease. The use of genetic engineering to identify one or more gene mutations (and combinations of

Weblink
Extension activity
Understanding cardiovascular disease through genome-wide association studies

these) in people with the disease allows the prediction of a predisposition or susceptibility to disease, essential for disease prevention.

Genetic studies have also led to improved screening, diagnosis and early intervention, essential to prevent non-infectious disease. Strategies to change behaviour, as well as planning and policies, can be put in place to try to prevent incidence of the disease.

Weblink
Genetic research in non-communicable disease in selected Arab countries

Genetic engineering to produce vaccines to prevent some cancers

Infection by the **human papilloma virus** (HPV) is linked to the majority of cases of cervical cancer and some anal, throat and other cancers. Genetic engineering techniques are used to produce vaccines such as Gardasil® and Cervarix®, which are effective in preventing infection by HPV. When administered before the age at which sexual activity begins, these vaccines are nearly 100% effective in preventing cervical and other associated cancers.

Newspix / Fiona Hamilton

FIGURE 17.14 The HPV vaccine prevents cancer by preventing infection by the human papilloma virus.

INVESTIGATION 17.2

A secondary-source investigation into prevention of non-infectious disease using genetic engineering

INTRODUCTION

In this investigation you will research information, summarise it and use it in a Socratic seminar or debate about the use of genetic engineering techniques to prevent non-infectious disease/disorders, and the ethical issues that need to be taken into consideration.

AIM

- To research further information about the use of genetic engineering to prevent non-infectious disease/disorders
- To evaluate the effectiveness of these methods in the prevention of non-infectious disease/disorders
- To use researched information to participate in a **Socratic seminar** or debate to discuss the benefits and risks of the use of genetic engineering to prevent disease, and the ethical issues involved

Critical and creative thinking

Ethical understanding

Information and communication technology capability

Literacy

METHOD

Working in teams, research a variety of reliable secondary sources to further investigate information about genetic engineering used to prevent non-infectious disease/disorders, to enable participation in a Socratic seminar (see weblink) or debate. In this seminar or debate, the benefits and risks associated with the use of genetic engineering methods to prevent disease will be discussed, including ethical considerations that need to be taken into account.

Personal and social capability

Weblink
How to run a Socratic seminar

Weblink
Socratic seminar II

Information to be researched should include:

1 further information about PGT, including:
- the numerous types of diseases that can be tested for
- whether gender selection is possible
- the costs involved
- whether there is any harm to the embryo
- the fate of rejected embryos
- who PGT is suitable for
- the benefits and risks
- any ethical considerations that need to be taken into account

2 further information about the prevention of nutritional disease by the creation and growth of transgenic crops that include essential nutrients, including:
- an update on the progress/use of golden rice
- further examples of the use of transgenic crops to prevent nutritional diseases. One suggestion is the research being carried out, particularly by the CSIRO, to genetically engineer canola plants to produce omega-3 long-chain fatty acids
- details about the transgenic crop, to describe how and why it is produced
- for each example, an evaluation of the ability of these transgenic crops to prevent particular nutritional diseases
- issues associated with the use of transgenic species in agriculture, including the benefits/risks, whether they are 'safe' to eat and their effects on 'wild' populations
- any ethical considerations that need to be taken into account

3 two other examples of the use of genetic engineering to prevent non-infectious diseases/disorders. Include the types of diseases they prevent, the methods used, ethical considerations and an evaluation of their effectiveness in preventing these diseases.

RESULTS

1 PGT:

a Address all points listed in the Method section for PGT.

b Create and complete a table like the one below, identifying the benefits and risks associated with the use of PGT.

BENEFITS	RISKS

c Outline any ethical issues involved with the use of PGT.

2 Transgenic crops to prevent nutritional diseases:

a Address all points listed in the Method section for transgenic crops. Use a table like the one below to collate your information for each example.

EXAMPLE OF TRANSGENIC CROP	OUTLINE OF FEATURES	WHERE/WHY IT IS USED	EVALUATION OF ITS EFFECTIVENESS IN PREVENTING NUTRITIONAL DISEASE

b Create a table like the one you used in Question **1b**, identifying the benefits and risks associated with the use of transgenic crops.

c Outline the ethical issues involved with the use of transgenic crops.

3 Use tables similar to those given above, to summarise information about other examples of the use of genetic engineering to prevent non-infectious diseases/disorders, an evaluation of their effectiveness in preventing the particular disease/disorder, the benefits and risks of the process and any ethical issues associated with the use of the process.

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DISCUSSION

Prepare a written discussion of the use of genetic engineering methods to prevent non-infectious disease/disorders. Your discussion should include the following:

- an outline of examples of the use of genetic engineering, along with an evaluation of their effectiveness in preventing disease
- a description of the benefits and risks associated with the use of each of the genetic engineering methods outlined
- an analysis of the ethical considerations associated with the use of genetic engineering methods to prevent non-infectious disease/disorders.

This written answer can be used where appropriate in the class Socratic seminar/debate.

CONCLUSION

Write a few summary sentences to address the aims of this investigation.

KEY CONCEPTS

- Genetic engineering can be used to prevent non-infectious disease/disorders but there are ethical issues associated with its use that need to be addressed.
- PGT involves testing 3-day-old embryos for specific gene mutations associated with a known disease/disorder before implantation.
- Transgenic crops are being developed to prevent nutritional disease.
- GWAS can be used to detect a predisposition to non-infectious disease with a genetic basis. Planning and policies can be put in place to try to prevent incidence of the disease.

CHECK YOUR UNDERSTANDING 17.2

1. Define 'genetic engineering'.
2. Outline the process of pre-implantation genetic testing (PGT) and the role it plays in preventing non-infectious disease.
3. How can transgenic crops be used to prevent nutritional disease?
4. Outline the ethical issues associated with the use of genetic engineering techniques to prevent disease.

People who have long-term infections of hepatitis B virus have a much greater risk of developing liver cancer. Administering the vaccine to prevent infection by the hepatitis B virus will lower the risk of developing liver cancer, for some people.

Developing strategies to prevent a non-infectious disease outbreak

Many types of diseases have caused concern regarding the health and wellbeing of the population of Australia and other countries, at various times. Public health authorities use epidemiological studies to monitor trends, in order to provide evidence of emerging patterns that indicate that a disease (such as lung cancer or melanoma) is becoming more prevalent in a population. This data can identify populations and geographic areas or locations at high risk. Planning and policies are then put in place to try to prevent further incidence of this disease. As a result of these formal initiatives, practical strategies are developed and put into place in the community to prevent, as much as possible, further increase in the incidence of the disease.

The strategies put in place depend on the characteristics of the specific disease but would include some form of education programs and campaigns. These programs would be distributed over many forms of media to raise awareness about the particular disease/disorder, its characteristics, its effects on the individual (both short- and long-term) and suggestions to

Worksheet
Prevention of an environmental disease by public health campaigns: skin cancer

Worksheet
Prevention of non-infectious diseases

FIGURE 17.15 One of the many Cancer Australia posts on the social media platform Twitter

prevent the development of the disease. Suggestions about ways to lessen the effects of the disease/disorder and advice about support services available would also be included in these campaigns.

Social media platforms such as Facebook, Twitter and Instagram are important ways in which information can influence many members of the population. These platforms can be used not only to disseminate the information but also in an interactive way, with real-time conversations in which questions can be answered.

Public health organisations can identify public health **influencers**, such as bloggers, who drive conversations about health topics. They can then develop a relationship with them so the influencers can drive conversations and get others to take notice of the topic. Celebrities who take up the cause are influencers who can have a dramatic effect. For example, the World Heart Foundation celebrates World Heart Day on 29 September each year. Last year, it encouraged people around the world to take 'Healthy Heart Selfies' and post them with hashtags on social media sites. Queen Latifah and her mother joined the cause, influenced by the American Heart Association, and posted a video about their experiences on YouTube and encouraged others to do the same. The involvement of this celebrity influencer had a massive effect on this campaign.

Legislation is often put in place by governments to protect the population by reducing exposure of the population to causative factors of the disease.

INVESTIGATION 17.3

A secondary-source and first-hand investigation to develop strategies to prevent a non-infectious disease outbreak

Critical and creative thinking

Information and technology communication capability

Literacy

Personal and social capability

INTRODUCTION

Preventing disease of any kind is much more desirable than treating/managing the disease. In this investigation, you and your team have been tasked with developing a series of strategies to prevent a non-infectious disease outbreak. You will use information from this text and your own researched information.

The non-infectious disease can be one of your choosing, or it can be a fictional scenario. You can further develop strategies that are already in place, or develop a set of strategies for a disease that has limited strategies in place. This will culminate in a display of your strategies, either to the class or to the school population in general.

Here are some suggestions:

- The prevalence of type 2 diabetes has increased markedly in the past ten years or more.
- Iodine deficiency is a nutritional disease that has become more prevalent in the Australian population.
- Many lifestyle diseases are easily preventable.

A fictional scenario you could use is described here, or you could make up your own.

In a rural community, the pesticides used on local farms have been distributed by wind to nearby townships, causing symptoms such as headaches, rashes, sore throats and stinging eyes in residents. Long-term exposure has been shown to cause severe and long-lasting symptoms, such as inflamed liver, difficulty breathing and inability to concentrate.

»

AIM

To develop strategies to prevent a non-infectious disease outbreak

METHOD

1. Working collaboratively, choose a non-infectious disease.
2. Research information from a wide variety of reliable sources about this disease, including: cause, symptoms, prevention.
3. Research data about the incidence, prevalence and mortality rate of the disease, to justify the implementation of strategies and to identify whether any groups/locations are more at risk than others (target groups).
4. Use this information to brainstorm ideas about strategies that could be implemented to prevent further incidence of the disease.

   These strategies should:

   - be directed towards the target group(s)
   - have a multifaceted approach that not only educates the public about the cause of the disease and the effects on the body but also provides guidance on preventative measures and support services available.

   The strategies could include, but are not limited to, the following (use your imagination to come up with new and interesting strategies):

   - catchy phrases, posters, advertising campaigns for print, radio and TV
   - a 'day' to raise awareness
   - images of the effects of the disease
   - changes in legislation that could be implemented to assist in prevention.
5. Design and document a plan that includes the following information:
   - the disease
   - cause, symptoms, methods of prevention
   - data to support the need for intervention strategies for prevention
   - identification of target groups
   - an overview of the variety of strategies that are to be developed
   - an explanation of how each strategy will assist in the prevention of your chosen disease
   - the expected outcome of the implementation of the strategies.
6. Further develop the strategies that have been outlined in the plan – for example, design and produce a poster, make a video, prepare a multimedia presentation, develop an attention-grabbing approach.
7. When you have finished developing your strategies, put together a portfolio of your strategies and display them, either in the classroom for peer review or for the school population in general.
8. Reference your sources in an acceptable format.

RESULTS

1. Present your results as a documented plan that includes all areas listed in the Method section.
2. Prepare a portfolio of strategies and put them on display.

DISCUSSION

1. Discuss the importance of disease prevention.
2. Why is it important to have a variety of strategies when trying to prevent a specific disease?
3. Explain how each of your strategies assists in the prevention of your chosen disease.
4. Discuss the role of epidemiology in developing these strategies.

CONCLUSION

Write a few summary sentences to address the aim of this investigation.

17 CHAPTER SUMMARY

Prevention: How can non-infectious diseases be prevented?

PREVENTION

Prevention

- Reduces suffering of individual, improves quality of life and overall health and wellbeing
- Reduces financial burden on individual and health system
- Reduces lifestyle diseases by changing people's behaviour

EDUCATIONAL/PUBLIC HEALTH PROGRAMS

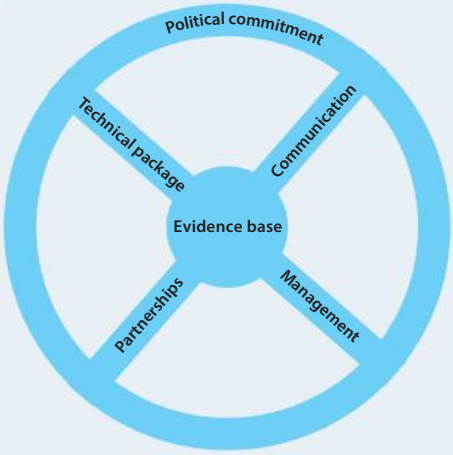
Epidemiological studies → Prevalent diseases, groups at risk

Strategies for changing behaviours (prevention) → Educational/public health programs, genetic engineering, government legislation

Educational/public health programs

National days, online resources, advertising campaigns to educate, posters, social media and influencers, apps, tailoring delivery of message, screening those at risk, national helplines, support programs, foundations.

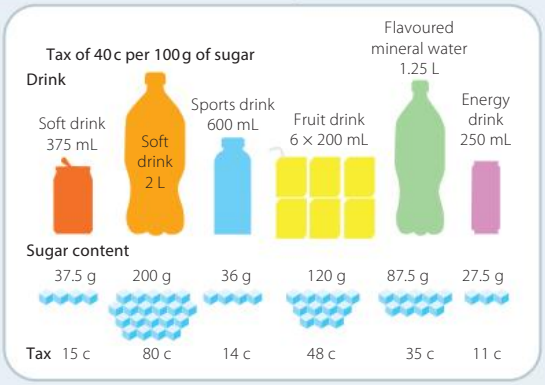
Examples: QUIT; Diabetes Helpline; Slip, Slop, Slap, Seek, Slide; Alcohol. Think again



LEGISLATION

Legislation aims to minimise the effects of risk factors in the population – tobacco, alcohol, unhealthy diet, lack of physical activity – by:

- prohibiting advertising and sponsorship
- increasing tax (levy) on risk factor
- restricting time and place for use of risk factor
- enforcing clear labelling of food.



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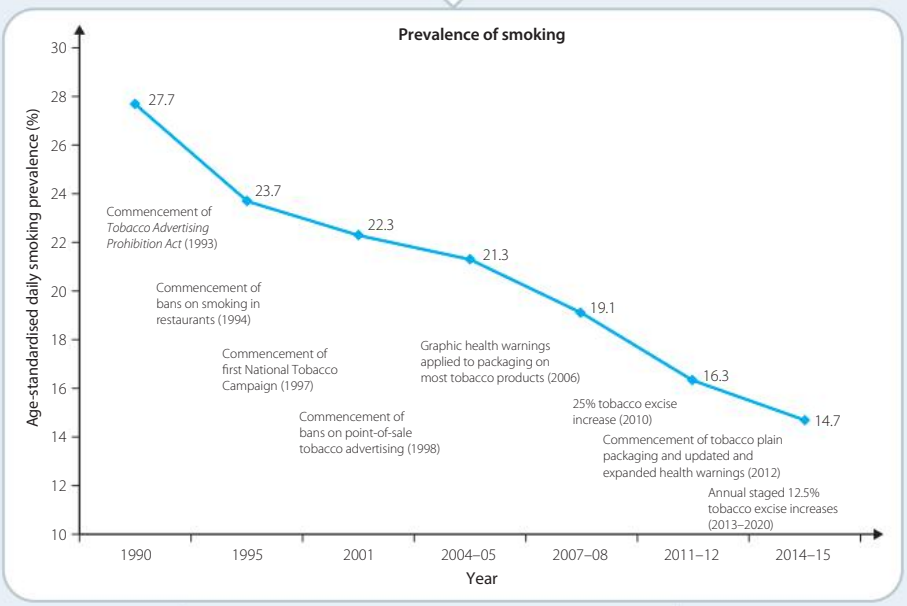
EVALUATION

Evaluate effectiveness of educational programs and campaigns – compare incidence and prevalence data before and after implementation of programs.

For example, QUIT campaign:

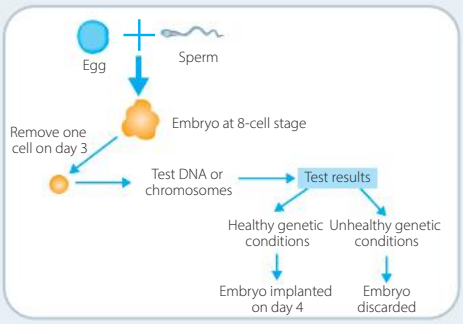
- decrease in smoking prevalence with implementation of strategies
- incidence and mortality rates of lung cancer – will eventually decrease for both males and females.



GENETIC ENGINEERING

Prevents:

- some genetic diseases, e.g. using pre-implantation testing and array comparative genomic hybridisation (aCGH)
- some cancers, e.g. production of vaccine to prevent cervical cancer
- some nutritional diseases, using transgenic species, e.g. golden rice.



Developing strategies to prevent a non-infectious disease outbreak

Epidemiological studies

↓

Populations and geographic areas at risk

↓

Planning and policies

↓

Strategies in place

↓

Prevention

17 CHAPTER REVIEW QUESTIONS

Review quiz

1 Discuss the benefits of disease prevention.

2 **a** Why are many non-infectious diseases/disorders considered preventable?

b Outline methods that could be used to change the behaviour of individuals in order to prevent lifestyle diseases/disorders.

3 Foetal alcohol spectrum disorder is a lifelong condition related to brain damage caused by foetal exposure to alcohol. Affected people may or may not have distinct facial features but all are characterised by cognitive, behavioural, health and learning difficulties. These difficulties are often characterised by lack of concentration, inability to follow cause-and-effect reasoning, impulsivity and language difficulties.

Develop three strategies that could be put in place to raise awareness of this condition and prevent its occurrence.

4 Exposure to silica dust increases the likelihood of developing lung cancer. Silica dust is made up of grains that are smaller than sand; it is formed when stone, rock, gravel, clay, bricks, concrete and tiles are cut. Tradespeople, home renovators, miners and other workers are at risk if this dust is inhaled.

Methods that involve raising awareness of the dangers of silica dust in training modules, as well as advertisements suggesting that PPE (personal protective equipment) be worn when cutting materials containing silica, have been developed. Outline how the effectiveness of these methods of prevention could be evaluated.

5 Assess the use of genetic engineering methods such as PGT and transgenic crops in the prevention of non-infectious disease/disorders. Include the ethical considerations that should be taken into account.

Exam preparation

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18 Technologies and disorders

INQUIRY QUESTION

How can technologies be used to assist people who experience disorders? CCT

Students:

- explain a range of causes of disorders by investigating the structures and functions of the relevant organs, for example:
  - hearing loss
  - visual disorders
  - loss of kidney function CCT
- investigate technologies that are used to assist with the effects of a disorder, including but not limited to: (ACSBL100) ICT L
  - hearing loss: cochlear implants, bone conduction implants, hearing aids ICT PS
  - visual disorders: spectacles, laser surgery ICT PS
  - loss of kidney function: dialysis PS
- evaluate the effectiveness of a technology that is used to manage and assist with the effects of a disorder (ACSBL100) EU L

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Assessments

- Chapter review
- Review quiz
- Exam preparation

Investigations

18.1 A first-hand and secondary-source investigation to study the structure and function of the ear

18.2 A secondary-source investigation into technologies to assist with the effects of hearing loss

18.3 A first-hand investigation to observe the structure of the eye

18.4 A first-hand investigation to model the process of accommodation

18.5 A secondary-source investigation of laser surgery in the correction of visual disorders

18.6 A first-hand investigation to identify the regions of the mammalian kidney

18.7 A secondary-source and first-hand investigation to research renal dialysis

18.8 A secondary-source investigation to evaluate the effectiveness of a technology

Worksheets

- Structure of the ear
- Structures in the brain responsible for auditory processing
- Find your blind spot
- Diagram and vocabulary of the eye

Nelson MindTap

To access these resources, visit **cengage.com.au/nelsonmindtap**

The human body is a complex integration of many systems that coordinate to form a functioning organism. It is astounding that so many complex 'parts' can all function properly, both separately and together as a whole. It is not surprising that at times certain parts may fail to function as required, leading to disorders. The characteristics of these disorders can usually be explained in relation to errors in the structure and/or functioning of a particular organ.

Progress in scientific research has led to the development of technologies that can be used to diagnose and correct many of these disorders, improving the quality of life of those who in earlier times would have been severely hindered by them.

FIGURE 18.1 Then and now: technology to diagnose and treat eye disorders has advanced remarkably since the early days of medicine.

18.1 The ear

The ear is a sense organ that provides a major communication pathway between the external environment and the body.

Structure and function of the ear

Our ability to hear properly relies on the complex structure and functioning of the ear, the **auditory nerve** and the brain.

The ear has evolved to transfer energy in different forms from the external environment to the brain. Sound waves (sound energy) are directed into the outer ear canal by the **pinna** and set up vibrations (kinetic energy) of identical frequency in the eardrum (**tympanic membrane**) (Fig. 18.2). The vibrations are transferred to tiny bones in the middle ear called the **ossicles** – the **hammer (malleus)**, **anvil (incus)** and **stirrup (stapes)**. These bones amplify and transfer kinetic energy to the **oval window**, which is the membrane that separates the middle ear from the fluid-filled inner ear. Vibrations pass from the oval window into the upper canal of the cochlea and then through the lower canal to the **round window**, which acts as a valve, relieving built-up pressure from the wave set up in the **perilymph** of the cochlea.

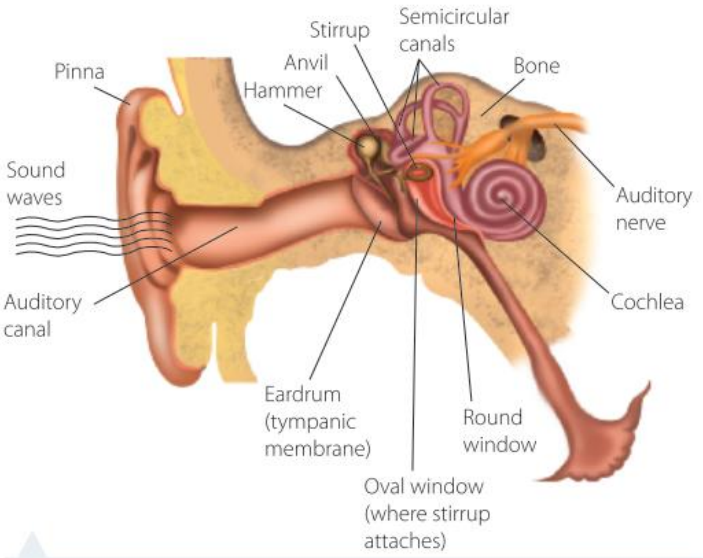
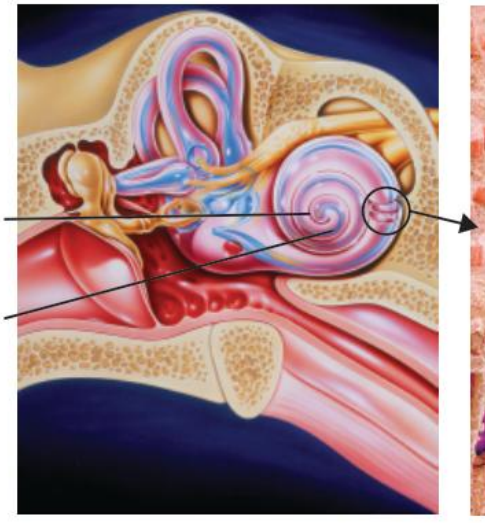


FIGURE 18.2 The structure of the outer, middle and inner ear

The fluid-filled cochlea is composed of three canals: upper, lower and middle (Fig. 18.3a). The middle canal contains the receptor cells in the **Organ of Corti**. The Organ of Corti sits on top of the **basilar membrane** and has an upper **tectorial membrane** and lower basilar membrane (Fig. 18.3b). Between these two membranes are **hair cells**, the receptor cells of the ear.

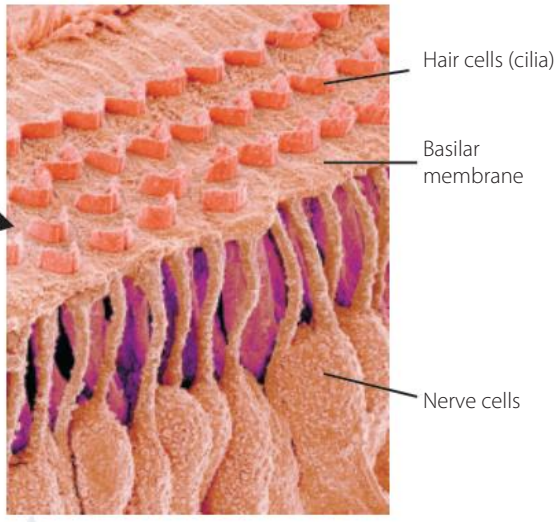
When a wave moves through the fluid-filled canals of the cochlea from the oval window, it pushes up on the basilar membrane (Fig. 18.4). This causes bending of the hair cells against the tectorial membrane, which is the stimulus that transforms the kinetic energy of the wave into the electrical energy of the nerve impulse sent along the auditory nerve to the brain.

Weblink
Pathway of sound through the ear



John Bavosi/Science Photo Library

FIGURE 18.3 The inner ear consists of fluid-filled passages made up of three canals: vestibular membrane, tectorial membrane and basilar membrane

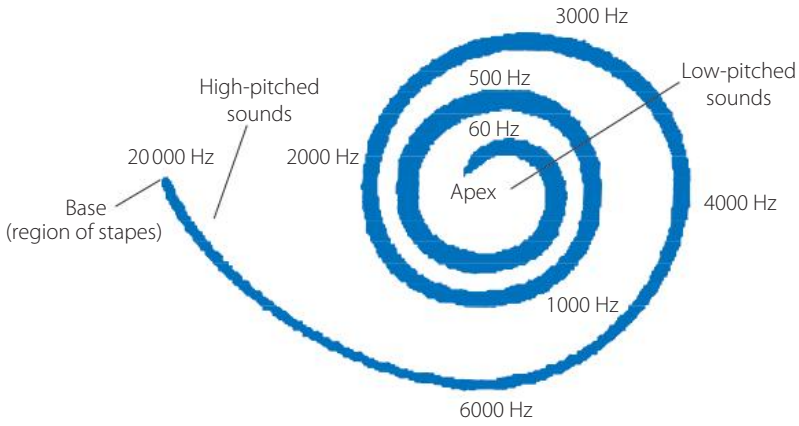


Susumu Nishinaga/Science Photo Library

FIGURE 18.4 An SEM of hair cells within the cochlea showing the cilia, which bend with sound triggering a neurochemical response that generates nerve impulses.

Sounds of different pitches are detected by hair cells at different regions of the organ of Corti (Fig. 18.5). High-pitched sounds stimulate hair cells closest to the oval window (base), while low-pitched sounds stimulate hair cells at the other end of the cochlea (apex). There is a finite number of hair cells, and if they are damaged, frequencies corresponding to the damaged cells will not be heard.

FIGURE 18.5
Vibrations of different frequencies stimulate hair cells at different positions along the cochlea.



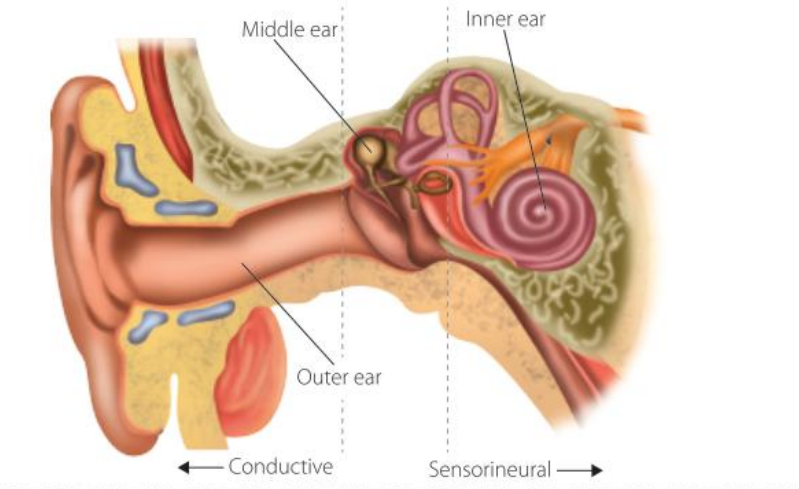
Hearing loss

Hearing loss can occur for various reasons, and the type of hearing loss depends on the particular area of the ear that has not been formed properly, has been damaged or is not functioning in the correct way.

There are two main types of hearing loss:

- conductive hearing loss
- sensorineural hearing loss (Fig. 18.6).

FIGURE 18.6
Incorrect transfer of vibrations in the outer and middle ear causes conductive hearing loss. Damage to the structures of the inner ear causes sensorineural hearing loss.



Conductive hearing loss

Conductive hearing loss is caused by a problem with the mechanical conduction of vibrations through the outer and middle ear. There are numerous causes of this type of hearing loss, including malformation of the structures in these regions of the ear, a perforated eardrum, infections in the outer/middle ear, damage to the ossicles due to trauma such as exposure to sudden loud noises (for example, explosions or head trauma), and hardening of the stapes bone. All these factors inhibit the

movement of vibrations through the outer and middle ear. It is the loudness of the sound that is usually affected in this type of hearing loss.

Sensorineural hearing loss

Sensorineural hearing loss is caused by damage to, or malformation of, the inner ear, including parts of the cochlea, the hair cells or the auditory nerve. The damage is usually permanent, as is the hearing loss, which affects the loudness and clarity of sound. The most common cause of sensorineural hearing loss is excessive noise exposure, while other causes include heredity, birth defects, infections, tumours, medication and ageing.

Damage to the inner ear (including the receptor hair cells) prevents the kinetic energy of the vibrations being transformed into electrical impulses to be sent to the brain for interpretation.

INVESTIGATION 18.1

A first-hand and secondary-source investigation to study the structure and function of the ear

Information and communication technology capability

Literacy

INTRODUCTION

In this investigation you will use physical and digital models to study the structure and function of the ear. You will also measure the loudness of sounds and determine the upper limit of loudness before damage occurs to the structures of the ear. Explanations of the causes of hearing loss will also be investigated.

AIM

To investigate the structure and functioning of the ear and explain causes of hearing loss

MATERIALS

Model of human ear
Sound meter

Worksheet
Structure of the ear

METHOD

1. Using the supplied model, identify the parts of the ear shown in Figure 18.2.
2. Label a diagram of the ear (see worksheet *Diagram of the ear*).
3. Research using the Internet to find a 3D interactive model of the ear. Describe your findings when manipulating the 3D models of the ear.
4. Construct a table to summarise the parts of the ear, and the structure and function of each part.
5. Interact with the weblink or similar to demonstrate that sounds of different frequencies are detected at different areas along the cochlea.

Weblink
Sound transduction in the human ear

6. Research a variety of sources to determine the region(s) of the brain responsible for the processing of information from the ear. Record this information on the worksheet *Structures in the brain responsible for auditory processing.*
7. Use a sound meter to record decibels of some common sounds. Research a variety of reliable sources to determine the decibel level of loud sounds such as a siren, cicadas, a plane taking off, fireworks, a 12-gauge shotgun firing, a rock concert. As part of this research, determine the upper level of loudness before damage occurs to structures within the ear.

   Summarise your findings in a table, beginning with the softest sound and ending with the loudest.
8. Use a variety of reliable sources to research different causes of hearing loss, and explain the cause of each type of hearing loss in terms of the structure and/or functioning of the different parts of the ear. Present your findings in a format agreed to with your teacher.

Worksheet
Structures in the brain responsible for auditory processing

»

DISCUSSION

1. Draw a flow chart to trace the pathway of sound waves through the structures of the ear.
2. How did using models of different types increase your understanding of the structure and function of the ear?
3. Discuss the importance of wearing protective devices when being exposed to loud noises.
4. Assess the relevance, reliability, accuracy and validity of your data sources.

CONCLUSION

Write summary sentences to address the aim of this investigation.

KEY CONCEPTS

- Problems with the structures and functioning of the ear can cause hearing loss.
- The pathway of the sound wave through the ear can be summarised as:
  Pinna → external auditory canal → tympanic membrane → hammer, anvil, stirrup → oval window → cochlea → round window
- The organ of Corti contains hair cells that are the receptors in the ear.
- Bending of the hair cells when a pressure wave pushes on the basilar membrane stimulates the formation of electrical impulses.
- Electrical impulses are transferred to the brain by the auditory nerve.
- Hair cells at the base of the cochlea detect the highest-pitched sounds, while hair cells at the apex of the cochlea detect the lowest-pitched sounds.
- Conductive hearing loss occurs when vibrations cannot be transferred effectively through the outer and middle ear.
- Sensorineural hearing loss occurs when the inner ear is damaged or malformed.

CHECK YOUR UNDERSTANDING 18.1a

1. Draw a schematic diagram of the structures that make up the ear. On this diagram, label the outer, middle and inner ear regions, and the individual structures within these, and draw arrows to indicate the pathway of sound waves through the ear.
2. Outline the function of the round window.
3. Which region(s) of the ear are:
   a. air filled
   b. fluid filled?
4. a. Identify the receptors in the ear.
   b. Where are these receptors located?
   c. What is their function?
   d. Describe how they are stimulated to produce electrical impulses.
5. Draw a diagram to represent where different frequencies of sounds are detected in the cochlea.
6. Identify the two types of hearing loss and their respective causes.
7. Outline the impact of damage to:
   a. the hair cells that are responsible for detecting vibrations of a high frequency.
   b. the eardrum or the ossicles.
8. Outline the effect of prolonged exposure to loud noise.

Technologies to assist with the effects of hearing loss

Hearing loss affects the quality of life and survival of many individuals. Scientific research into technology that can be used to assist those suffering from hearing loss has led to the development of many ways to counter its effects. The specific technology used to assist with the effects of hearing loss depends on the cause of the hearing loss.

Conductive hearing loss can be assisted with the use of hearing aids and bone conduction implants, while sensorineural hearing loss can be assisted with the use of both hearing aids and cochlear implants.

Hearing aids are used to magnify the sound vibrations to better enable their transmission to the middle ear and then the inner ear.

A bone conduction implant has a microphone that detects the sound and transforms it into vibrations, which are then passed via the implant to the bone above the ear. These vibrations are directed through the bone to the cochlea, where they are processed as normal. These implants bypass the malformed or damaged outer and middle ear and transfer the vibrations directly to the inner ear.

A cochlear implant uses an external speech processor and transmitter coil. Internally, a receiver is attached to an electrode array implanted in the cochlea. The microphone of the speech processor detects sound, which is converted into a digital signal that is sent to the transmitter, then to the receiver. The digital signals are converted to electrical signals, which are then sent to the electrode array in the cochlea, where nerve endings are stimulated. These signals are then sent to the brain for processing.

Weblink
Hearing loss and hearing aids

Weblink
Bone conduction implants
Summarise the process of bone conduction implants to assist in hearing loss.

Weblink
Cochlear implants
Watch the animation and draw a flowchart to show the steps involved in hearing using a cochlear implant.

INVESTIGATION 18.2

A secondary-source investigation into technologies to assist with the effects of hearing loss

INTRODUCTION

In this investigation you will research information about technologies that assist with hearing loss: hearing aids, bone conduction implants and cochlear implants.

Information and communication technology capability

Personal and social capability

AIM

To investigate technologies that assist with hearing loss

METHOD

1. Work in pairs or groups to research information from a variety of reliable sources about the following technologies that assist with the effects of hearing loss: hearing aids, bone conduction implants and cochlear implants.
2. For each type of technology:
   - **a** find information about the type of hearing loss the technology can assist with, including suitable candidates
   - **b** provide a description of the technology, where it is positioned and how it assists with hearing loss
   - **c** provide a labelled diagram of the technology and digital references/videos to explain its use
   - **d** describe the benefits of the technology
   - **e** describe the limitations of the technology.
3. Information about each type of technology should be presented in a manner of your choosing and be presented to the class for peer review.

»

» DISCUSSION

1 Discuss the impact of the technologies available to assist with hearing loss on:
   a the lives of people who have hearing loss
   b the burden of hearing loss on the health system
   c the families of those suffering from hearing loss.
2 Assess the relevance, reliability, validity and accuracy of the information you have collected.
3 Suggest other characteristics of the technologies that could have been researched.

CONCLUSION

Write a few summary sentences related to the aim of this investigation.

KEY CONCEPTS

- In cases of conductive hearing loss, hearing can be assisted by the use of hearing aids and bone conduction implants.
- Hearing aids magnify the sound waves to assist their passage through the outer and middle ear to reach the inner ear.
- Bone conduction implants bypass the outer and middle ear by sending the vibrations through the bone above the ear straight to the inner ear.
- Hearing aids and cochlear implants assist hearing in cases of sensorineural hearing loss.
- Cochlear implants involve the conversion of sound into electrical impulses that stimulate an electrode array implanted in the cochlea, which in turn stimulates electrochemical impulses in the auditory nerve.

CHECK YOUR UNDERSTANDING 18.1b

1 a Identify two technologies that assist with sending vibrations to the inner ear when there are problems with the outer and/or middle ear.
   b Explain how each of these technologies functions.
   c What are the advantages and disadvantages associated with the use of each of these technologies?
2 a Outline the conditions under which a cochlear implant is used to assist a person who has hearing loss.
   b Draw a labelled diagram to show the components of cochlear implant technology and describe how it works.
   c Discuss the advantages and disadvantages of cochlear implants.

18.2 The eye

The eye is a complex structure that contains receptors, called **photoreceptors**, that receive light energy from the external environment and convert it to nerve impulses (electrical energy), which are sent via the **optic nerve** to the brain, where they are interpreted as images.

Structure and function of the eye

The eye is composed of three layers (Fig. 18.7).

- The **sclera** forms the opaque, tough protective coat that surrounds the majority of the eye, with the front section forming the transparent, curved **cornea**.
- The middle layer of the eye is the **choroid** layer and contains most of the blood vessels. The posterior of the choroid layer is black, to make the eye 'light tight' and reduce scattering or reflection of light in the eye. The forward section of the choroid layer forms the **ciliary body**, the **lens** and the **iris**.

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- The inner layer of the eye is made up of the **retina**.

Light receptors are located on the retina, which covers the rear portion of the eye. In order to form clear, focused images, these light rays must be refracted (bent) so that a sharp focus is formed on the retina (Fig. 18.8). The structures within the eye that the light passes through allow the correct degree of refraction to occur when all parts are functioning correctly.

The reflected light from an object in the environment passes into the eye through the cornea. The curvature of the cornea refracts or bends the incoming light. It then passes through fluid called the **aqueous humour**, situated between the cornea and the lens. This liquid further refracts the light to a small degree. The light travels through the **pupil** – the area in the centre of the eyeball that appears black. The pupil is a 'hole' that can be made larger or smaller by a ring of muscle called the iris (the coloured part of the eye). When the light intensity is high, the iris expands to make the pupil smaller, thus restricting the amount of light entering the eye. The opposite occurs when the light is dim, to allow more light into the eye.

The light moves through the pupil and through the lens, a highly elastic transparent biconvex structure enclosed within the **lens capsule**. The curvature of the lens is changed by the ciliary muscle, so the lens can refract the incoming light to form a focused image on the retina. This allows focused vision of objects at different distances. The process by which the lens changes curvature is called **accommodation**.

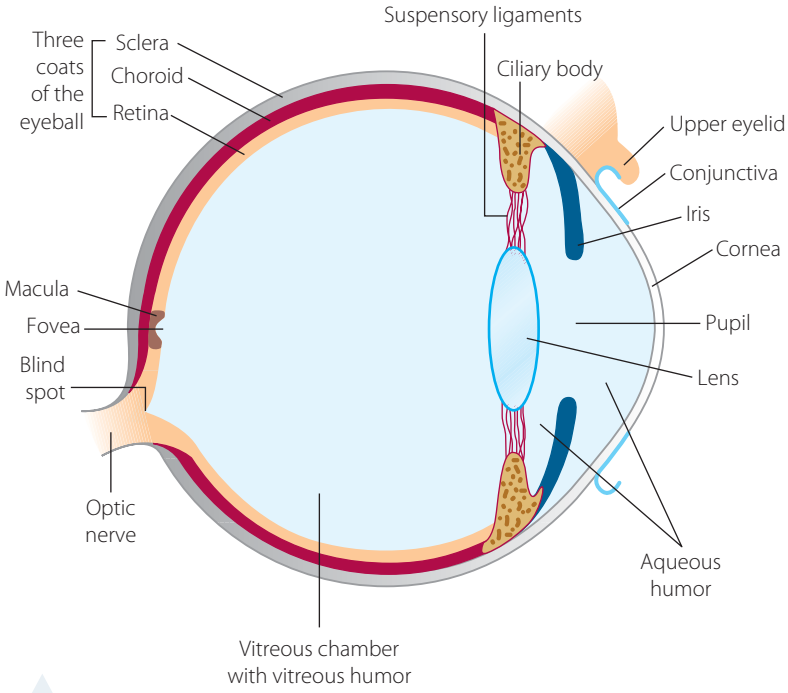

FIGURE 18.7 Structure of the mammalian eye

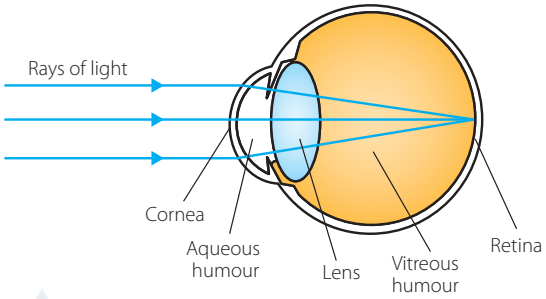

FIGURE 18.8 Refraction of light rays as they pass through the eye to form a sharp focus on the retina

Accommodation is described in more detail on pages 601–2.

Rods and cones are discussed in more detail on pages 605–6.

Once the refracted light passes through the lens, it moves through the **vitreous humour** to the retina. The vitreous humour is a jelly-like clear fluid that also refracts the light and helps to maintain the shape of the eyeball.

The retina covers the posterior two-thirds of the eyeball and is extremely thin and delicate. It contains several layers of nerve cells, including two types of photoreceptor cells, **rods** and **cones**, that are situated closest to the back of the eye. The rod and cone cells are stimulated by light focused on the retinal surface, and transform this light energy into electrical nerve impulses that are sent via the optic nerve to the brain.

A small section of the retina, the **fovea** (Fig. 18.7), is the area of greatest **visual acuity** (the ability to see a clear and precise image). This area is a depression in the **macula** and is directly opposite the incoming light. It contains many densely packed cone cells but no rod cells.

The area at the back of the eye, where the nerve fibres leave the eye and converge to form the optic nerve, is called the **blind spot**. Light that falls here is not detected, as there are no photoreceptors. Instead, the brain fills in the missing parts, so you see a whole image.

Weblink
Structure and function of the eye
Summarise the structure and functioning of the eye

Worksheet
Find your blind spot

INVESTIGATION 18.3

A first-hand investigation to observe the structure of the eye

Weblink
Virtual eye dissection

INTRODUCTION

In this investigation, the structure of the eye will be investigated by dissection and the use of models, both digital and physical. If an actual dissection is not performed, there are a number of online interactive, virtual dissections that can be used (see weblink).

AIM

To investigate the structure of the eye and relate the structures to their functions

RISK ASSESSMENT

RISK ASSESSMENT

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?
Scalpel/scissors	Sharp edges can cause cuts	Use scalpel/scissors with care, hold by the handle and keep fingers away from sharp edge of scalpel/scissors.
Use of animal material	Poisoning if ingested or enters through skin	Wear gloves at all times. Wash hands at conclusion of investigation. Dispose of waste material in an acceptable fashion.

What other risks are associated with your investigation, and how can you manage them?

MATERIALS

- sheep/cow/pig eyeball
- scalpel
- forceps
- scissors
- gloves
- dissecting tray
- newspaper
- model of eye
- image of eye with structures labelled
- digital model of eye
- blindfold

METHOD

1. Work with a partner. One student is to put on the blindfold and be guided by the other student as they try to navigate their way around the classroom, hallway or school grounds. Roles should be swapped to give each student the opportunity to experience activities without the benefit of sight.
2. Observe the different structures of the eye using the models and diagrams supplied.
3. Research a variety of reliable sources to determine the region of the brain that receives information for processing from the optic nerve, and shade this area on a diagram of the brain from Investigation 18.1.
4. Obtain the equipment for dissection of the eyeball, or follow an online virtual dissection.
5. Identify the external structures of the eyeball.
6. Cut off all the fat and muscle attached to the eyeball, taking care not to cut off the optic nerve protruding from the back.
7. Roll the eyeball between your hands for 3–5 minutes to loosen the tough sclera.

8 Using forceps, pinch an area of the sclera about halfway between the front and back of the eye. Using the scissors, cut through the sclera in the pinched area.

9 Place the scissors into this small hole with the tips pointing upwards and cut around the eyeball to separate the front half from the back half (Fig. 18.9).

10 Peel the front half of the sclera away and hold it up to the light. Observe the iris and the 'hole' that is the pupil.

11 Using your hands and fingers, gently peel the back of the eye away from the vitreous humour without 'bursting' it.

12 Gently pry the lens away from the vitreous humour and place it on the newspaper. What do you see?

13 Observe the layers within the inside of the back section of the eye, noting any distinctive features and the place where the optic nerve leaves the eye.

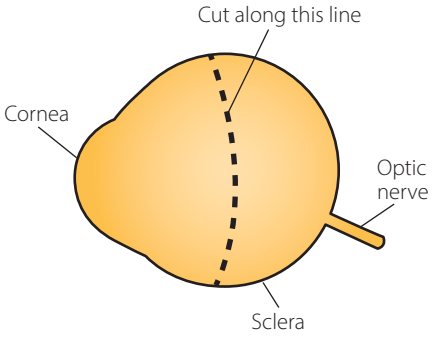

FIGURE 18.9 External structure of the eyeball, showing the dissection line

14 The retina is a delicate beige or grey layer that is very thin and may be hanging off the edges of the optic nerve. The choroid layer is the thin black layer below the retina. The choroid layer of a cow eye consists of a coloured layer that is not present in human eyes.

RESULTS

1 Describe what it was like to negotiate the classroom/hallways/school grounds without the ability to see.

2 Label the structures indicated on the worksheet *Diagram of the eye*.

3 On the brain diagram from Investigation 18.1 (page 595), shade the areas where the messages from the eye are processed.

4 Draw a labelled diagram of the external structure of the eyeball.

5 Describe the structure of the sclera, the iris and the pupil.

6 Draw a labelled diagram of the vitreous humour with the lens attached.

7 Describe the structure of the lens and what you noticed when you placed the lens onto the newsprint.

8 Outline the structures you observed on the back portion of the eye: the retina, the choroid layer and the optic nerve.

Worksheet
Diagram of the eye

DISCUSSION

1 Prepare a table to summarise the different structures that are part of the eye, including a description of each structure and its function.

2 Describe how the dissection, use of models and being blindfolded increased your understanding of the structure and function of the eye.

CONCLUSION

Write summary sentences to address the aim of this investigation

Accommodation

In order to form a sharply focused image, on the retina, of objects at any distance, the lens must change its curvature to refract the light by the correct amount. Light entering the eye from close objects needs to be refracted more than light coming from distant objects. The greater the curvature of the lens, the more the light passing through it is refracted (greater refractive power).

Because the lens of the eye has elasticity, its curvature can be changed by the actions of the ciliary muscles and suspensory ligaments. When the eye views distant objects, the ciliary muscles relax, drawing the sclera back, which holds the **suspensory ligaments** taut (Fig. 18.10a). This has the effect of lengthening the lens, decreasing the curvature and reducing the refraction of the light.

To focus a clear image of a close object on the retina, the curvature of the lens must be increased, to increase the refractive power of the lens. This is achieved by the ciliary muscles contracting, which draws the sclera forward and releases the tension on the suspensory ligaments (Fig. 18.10b). This allows the lens to become rounder, increasing the curvature, and refracting the light to a greater degree.

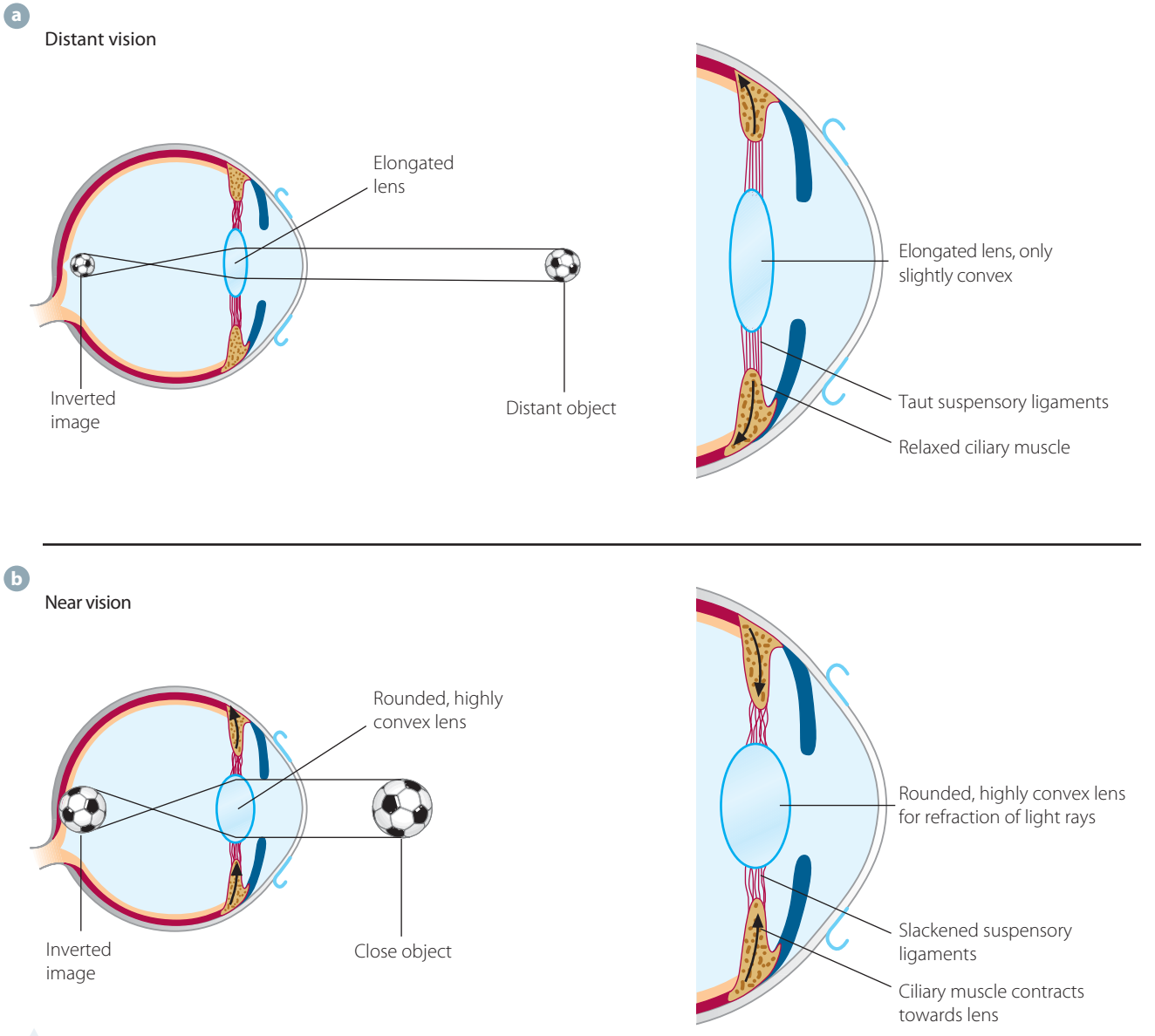

FIGURE 18.10 Accommodation by the lens, for **a** distant and **b** near vision

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INVESTIGATION 18.4

A first-hand investigation to model the process of accommodation

INTRODUCTION

Models are useful because they can make a concept simpler and assist in the understanding of more difficult concepts. In this investigation, the process of accommodation will be modelled to assist in the understanding of how lenses of different convexities can focus images from different distances.

RISK ASSESSMENT

WHAT ARE THE HAZARDS?	WHAT RISK DOES THIS HAZARD POSE?	HOW CAN YOU SAFELY MANAGE THIS RISK?
Glass lenses	Sharp edges can cause cuts	Use lenses with care. Don't drop them or they will break and create sharp edges
Candles/matches	Burns to the skin	Use candles and matches with care and keep body parts away from burning flames

RISK ASSESSMENT

What other risks are associated with your investigation, and how can you manage them?

MATERIALS

- lens holder
- metre rule
- clipboard
- blank paper
- candle
- matches
- biconvex lenses of different convexities

METHOD

1. Set up the equipment as shown in Figure 18.11.
2. Keep distance A the same at all times.
3. Identify what the parts of the model correspond to in the actual process of accommodation.
4. Place one of the lenses in the lens holder and move the candle slowly either towards or away from the lens until a clear image of the flame is focused on the blank paper.
5. Measure distance B.
6. Replace the lens with a lens of a different convexity and note whether the flame is still in sharp focus.
7. Again move the candle until it is in sharp focus and measure distance B.
8. Repeat steps 6 and 7 with a third lens, of a different convexity.

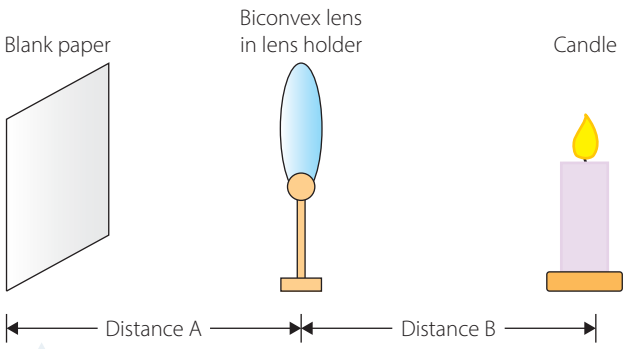


FIGURE 18.11 Equipment used to model the process of accommodation

RESULTS

1 Copy and complete the following table.

LENS CONVEXITY	DISTANCE B* (cm)
Most convex	
Mid convexity	
Least convex	

*distance of candle from the lens

2 Present the results obtained above in a different form.

3 Copy and complete the table below to identify what the parts of the model represent in the actual process of accommodation

ACTUAL PROCESS OF ACCOMMODATION	PART OF THE MODEL
Distance between the lens and the retina	
	Glass lenses of different convexity
	Candle
	Distance between candle and lens

DISCUSSION

1 In the model used, identify the:
   - **a** independent variable
   - **b** dependent variable
   - **c** controlled variables.

2 How did the distance between the candle and the lens vary in relation to the convexity of the lens used?

3 Outline how the results of this investigation demonstrated the process of accommodation.

4 Discuss the validity of this model.

5 **a** Outline the limitations of this model of accommodation.
   b Suggest improvements that could be made to this investigation.

6 How was your understanding of the process of accommodation assisted by this investigation?

CONCLUSION

Write a few summary sentences to address the aim of this investigation.

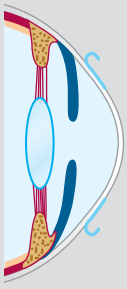
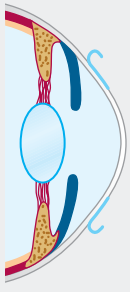
KEY CONCEPTS

- The pathway of light through the eye is summarised as:
  Cornea → aqueous humour → pupil → lens → vitreous humour → retina
- A focused image is formed on the retina by the refraction of incoming light rays by the cornea, the aqueous humour, the lens and the vitreous humour.
- Light enters the eye through the pupil. The iris controls the size of the pupil, which in turn controls the amount of light entering the eye.
- The process by which the lens changes its curvature to focus on objects at different distances is called accommodation.
- To focus on close objects, the light has to be refracted by a large amount, so the lens has the greatest curvature. This is achieved when the ciliary muscles contract and move the sclera forward, loosening the suspensory ligaments.
- To focus on distant objects, the lens must be relatively flat, as less refraction is required. This is achieved when the ciliary muscles relax, drawing the sclera back and causing the suspensory ligaments to become taut, pulling the lens into a long, flat shape.

- The retina lines the back of the eye and contains the receptors, rods and cones, which detect light rays and convert them into electrical impulses to be sent to the brain via the optic nerve.
- The fovea is an area on the retina directly opposite the incoming light that contains only cones and is the region of greatest visual acuity.
- The tough outer sclera protects the eye and continues to form the clear, transparent cornea.
- The choroid layer is mostly black, to make the eye 'light tight'.

CHECK YOUR UNDERSTANDING
18.2a

1 Draw a schematic diagram of the eye and label the components as shown in Figure 18.7.
2 Outline the structures through which the light passes to be refracted to form a focused image on the retina.
3 Define 'accommodation'.
4 Copy and complete the following table to summarise the process of accommodation

TYPE OF VISION	SHAPE OF LENS	ACTION OF MUSCLES OF CILIARY BODY	TENSION OF SUSPENSORY LIGAMENT	DIAGRAM OF ANTERIOR PART OF EYE, SHOWING SHAPE OF LENS
Distant vision				
Near vision				

5 **a** Summarise the procedure used to model the process of accommodation, identifying how the different components of the model represent the structures present in the eye.
b Relate this model to the process of accommodation.

Photoreceptor cells: rods and cones

Rods and cones are the receptor cells of the eye. They are located in the retina, where they receive focused light energy and convert it into electrochemical impulses that are sent to the brain for interpretation as images.

In humans, each retina contains approximately 125 million rod cells and 6–7 million cone cells. Rods are evenly distributed over most of the retina, but are absent from the fovea. Cones are distributed in groups throughout the retina, but are lower in number on the periphery. They are most concentrated in the fovea.

Both rods and cones have a similar structure, consisting of elongated cells that contain an **outer segment** joined to an **inner segment** that leads to the conducting part of the cell. This part of the cell comprises a cell body containing the nucleus, and an extension process called the foot, which conducts the nerve impulses to the next layer of neurons in the retina.

Rods have a long, narrow rod-shaped outer segment, while cones have a shorter, thicker cone-shaped outer segment (Fig. 18.12).

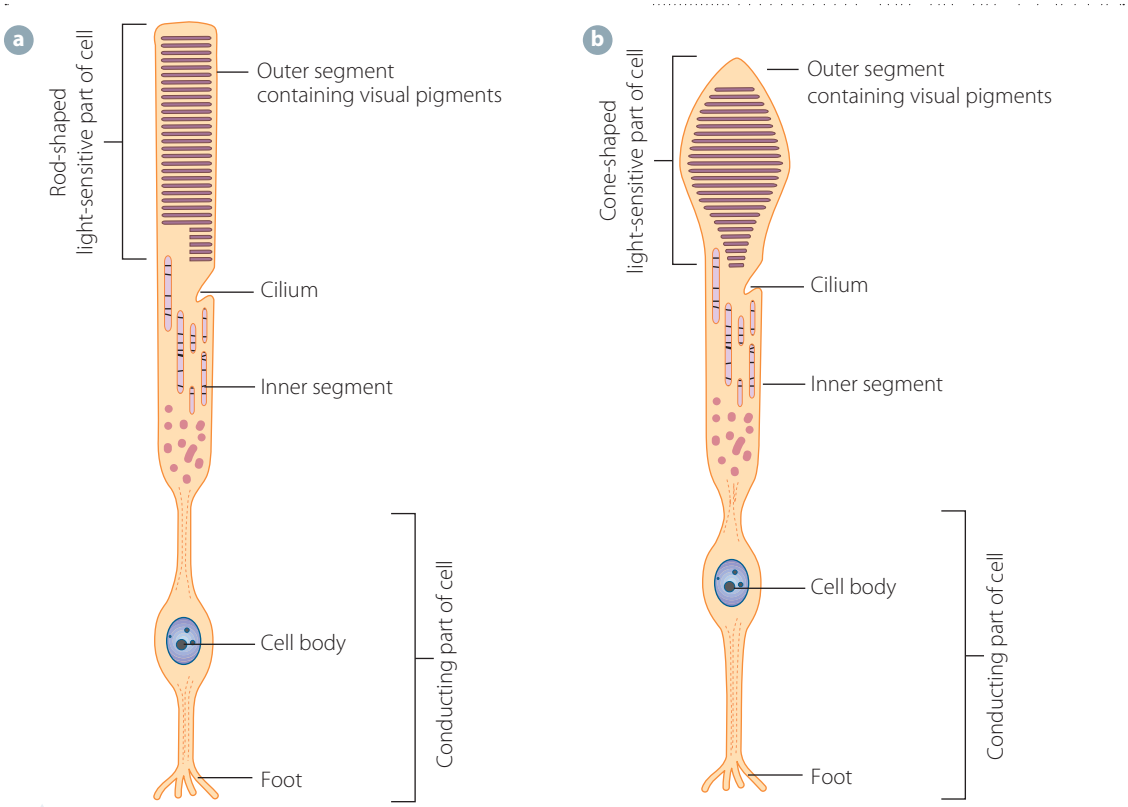


FIGURE 18.12 The structure of **a** a rod cell and **b** a cone cell

Both rods and cones contain chemical substances, called visual pigments, stacked in layers in their outer segments. The role of these visual pigments is to absorb light energy and convert it into electrochemical impulses for the brain to interpret.

Rods contain only one type of pigment, called **rhodopsin**, and cannot detect colour. They are extremely sensitive to low levels of light and are responsible for night vision, detection of light and shadow contrasts and movement, as well as peripheral vision.

Each cone contains one of three types of visual pigments, called **iodopsins**. Each type of iodopsin is sensitive to a different wavelength of light: red, green or blue. Cones are responsible for colour vision, and it is believed that all colours perceived by the eyes are a combination of the wavelengths detected by the different types of cones. Each iodopsin pigment has a wavelength for peak sensitivity, and can also detect light on either side of these peaks, which overlaps with the other iodopsins. Therefore, light of one particular wavelength may stimulate more than one type of cone. By comparing the rate at which various receptors respond, as well as the overlap in colours detected, the brain is able to interpret these signals as intermediate colours.

Weblink
Photoreceptors

Worksheet
Vocabulary of the eye

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The light-sensitive pigments, rhodopsin and iodopsin, have a similar chemical structure and each is made up of two parts:

- a **retinal** (**retinene**) molecule that is derived from vitamin A
- a protein, called **opsin**.

The type of opsin present depends on whether the visual pigment is rhodopsin or iodopsin.

When rods and cones are exposed to light, it causes the retinal molecule to change its form (Fig. 18.13). This initiates a reaction that causes the light energy to be transformed into an electrochemical impulse, which moves along the neurons to the brain. At the same time, the opsin and retinal molecule dissociate and the receptor is '**bleached**'.

After a time, the two parts of the pigment re-join and are able to receive more light stimuli.

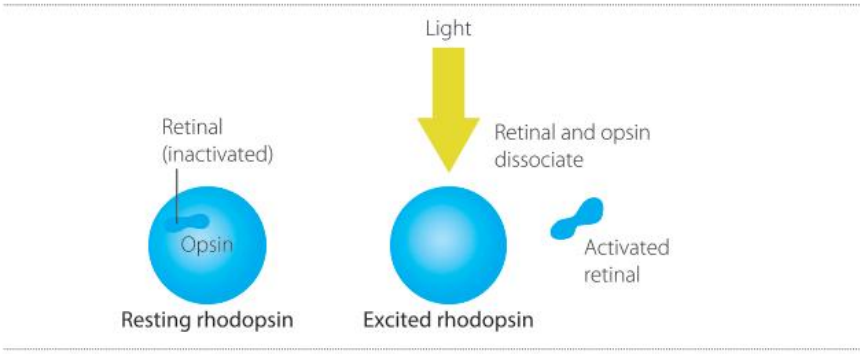

FIGURE 18.13 Excitation of rhodopsin by light

Visual disorders

The ability of the eye to function effectively to allow 'normal' vision depends on the individual components of the eye having the correct structure and function.

Long- and short-sightedness

The most common types of visual disorders are **myopia** (short-sightedness) and **hyperopia** (long-sightedness).

A person who has *myopia* can see near objects clearly, but distant objects appear blurred. This occurs because the focused image from a distant object falls in front of the retina, while the image from a near object is focused onto the retina (Fig. 18.14a).

Myopia can have a number of causes:

- The shape of the eyeball may be too elongated.
- The refractive power of the cornea may be too strong.
- The lens may not flatten enough when the ciliary muscles contract.

A person who has *hyperopia* can see distant objects clearly, but near objects are out of focus. This is because the focused image would fall behind the retina (Fig. 18.14b).

Possible causes of hyperopia:

- The eyeball is too rounded – that is, too short from front to back.
- The lens is too flat and is unable to achieve the required convexity. This could be due to loss of elasticity in old age.
- The refractive power of the cornea is too weak for the shape of the eye.

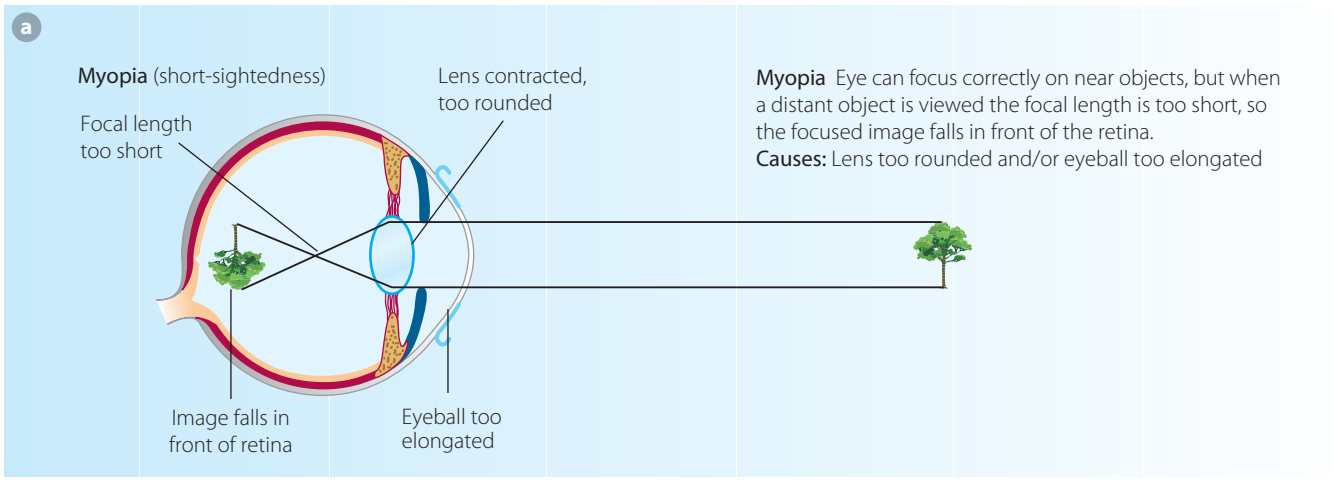

FIGURE 18.14 Two common visual disorders: **a** myopia (short-sightedness) and **b** hyperopia (long-sightedness)

Cataracts

A **cataract** is the clouding of the lens (Fig. 18.15), which reduces the transmission of the light through the lens. This causes blurred vision of both near and far objects, and increased sensitivity to the glare of bright sunlight.

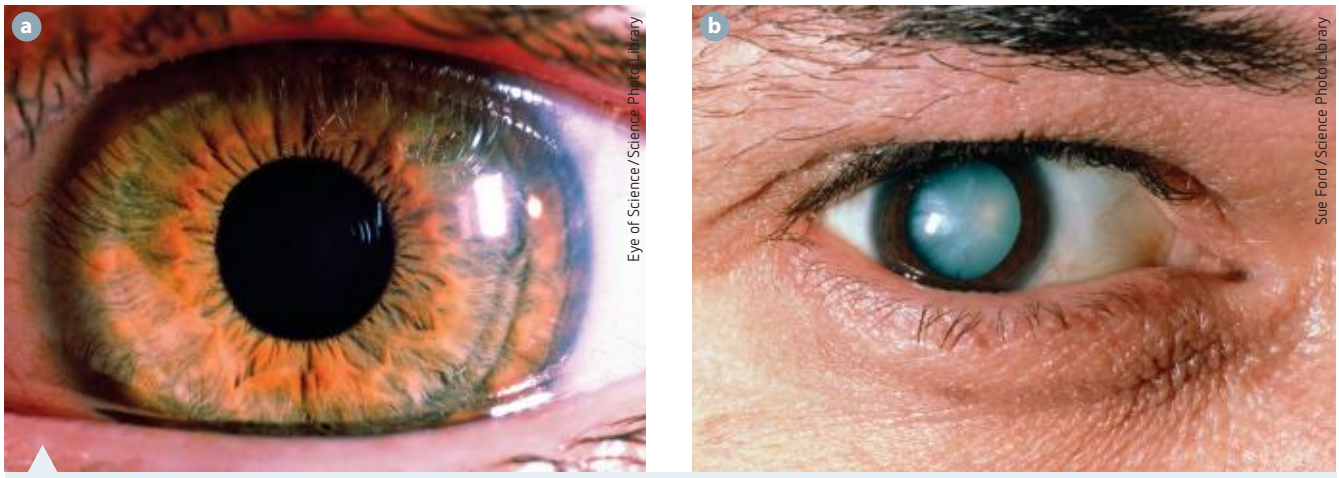

FIGURE 18.15 Cataracts cause visual disorders due to clouding of the lens: **a** a normal lens; **b** a lens with cataract

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Macular degeneration

Macular degeneration is a disease that causes degeneration of the cells beneath the retina, called the retinal pigment epithelium. This disease is responsible for 50% of all cases of blindness and is the leading cause of legal blindness in Australia. Central vision associated with the macula is affected (Fig. 18.16); peripheral vision is unaffected. Macular degeneration affects the ability to read, recognise faces, drive and carry out everyday tasks.

Degeneration of retinal pigment epithelial cells prevents light from being focused successfully on this area, leading to a loss of vision. Early detection is essential to slow the progression of the disease. Many **optometrists** now offer this check as part of a regular eye check. The risk of developing the disease can be reduced with lifestyle changes, including not smoking, following a healthy, balanced diet, doing regular exercise and reducing one's exposure to UV light.

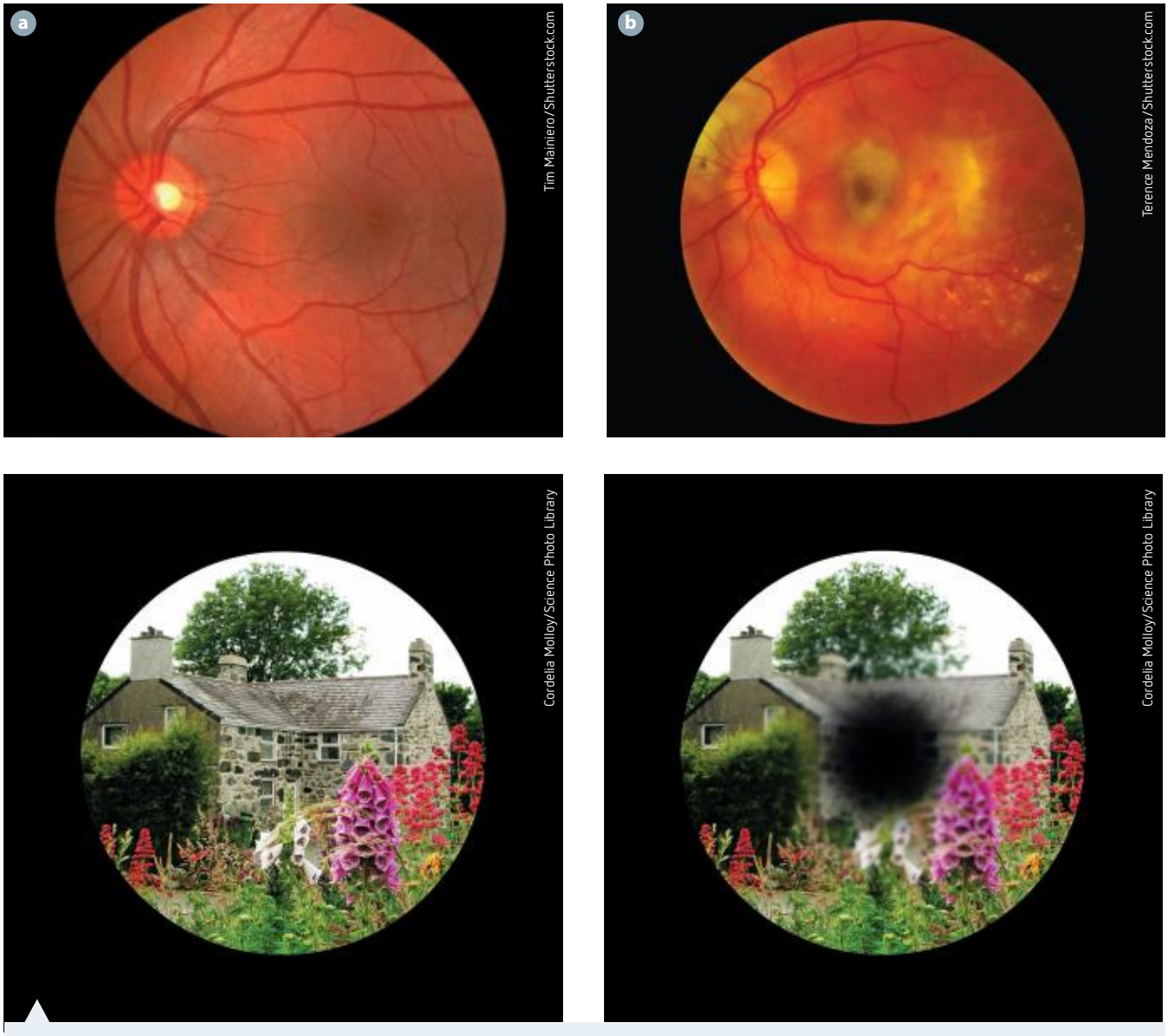


FIGURE 18.16 **a** A healthy macula and resulting image; **b** macular degeneration and resulting image

KEY CONCEPTS

- Rods and cones are the receptor cells in the retina of the eye. The visual pigments present in each of these cells change light energy into electrochemical impulses to be sent to the brain for processing.
- Rods contain rhodopsin and are responsible for the detection of light and vision in low light.
- Each cone contains one of three iodopsin pigments (blue, green and red). Cones are responsible for colour vision. The multitude of colours detected is due to the combination of each of the cones.
- All pigments are composed of retinal (a derivative of vitamin A) and an opsin (a protein). The type of opsin depends on the type of pigment.
- The detection of light by the pigments stimulates the formation of an electrochemical impulse and the splitting of the retinal and opsin. When the two parts of the molecule recombine, the pigment is ready to receive more light energy.
- Rods are distributed all over the retina.
- Cones are in clumps, with fewer on the edges of the retina.
- Common visual disorders such as myopia and hyperopia are caused by refractive errors of the eye.
- Cataracts are caused by clouding of the lens.
- Macular degeneration is caused by degeneration of the cells in the layer beneath the retina.

CHECK YOUR UNDERSTANDING 18.2b

1 Copy and complete the following table to summarise the characteristics of photoreceptors in the eye.

PHOTO-RECEPTOR CELL	DISTRIBUTION	STRUCTURE (DESCRIPTION AND DIAGRAM)	PIGMENT	FUNCTION	COLOUR(S) TO WHICH SENSITIVE
Rods					
Cones					

2 Outline the process by which light energy is converted into electrochemical impulses.

3 **a** Define 'myopia' and 'hyperopia'.

b Explain the cause of each of these refractive disorders of the eye. Draw diagrams to aid your explanations.

4 Describe the cause of each of the following, in terms of the structure and function of the eye:

a cataracts

b macular degeneration.

Technologies to assist with visual disorders

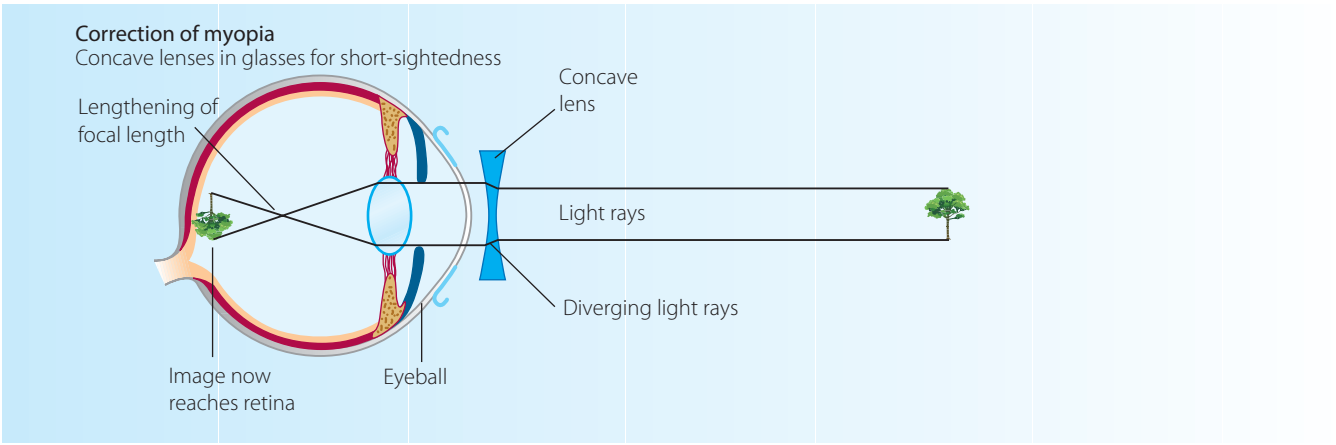
Visual disorders such as myopia and hyperopia cause distinct disadvantages to sufferers if they are not corrected. Myopic people have trouble seeing traffic signs, watching television or movies, recognising others, and pursuing leisure activities, including many types of sport. People with hyperopia have difficulty reading books, computer or phone screens, manipulating machinery and tools, and carrying out any activity that involves close-up hand–eye coordination. Technologies to assist with these and other visual defects are therefore an advantage to both the individual and society as a whole.

Spectacles

Spectacles (glasses) are frames that hold corrective lenses made of clear, hard plastic. The shape of the lens used is determined by the visual disorder that needs correction.

Myopia can be corrected by wearing spectacles with **concave lenses**, which are thicker towards the outside and thinner towards the centre. These lenses bend the light rays outwards, causing them to diverge before they reach the eye. This extends the focal length of the light rays, allowing the focused image of a distant object to fall on the retina instead of in front of it (Fig. 18.17a).

a



b

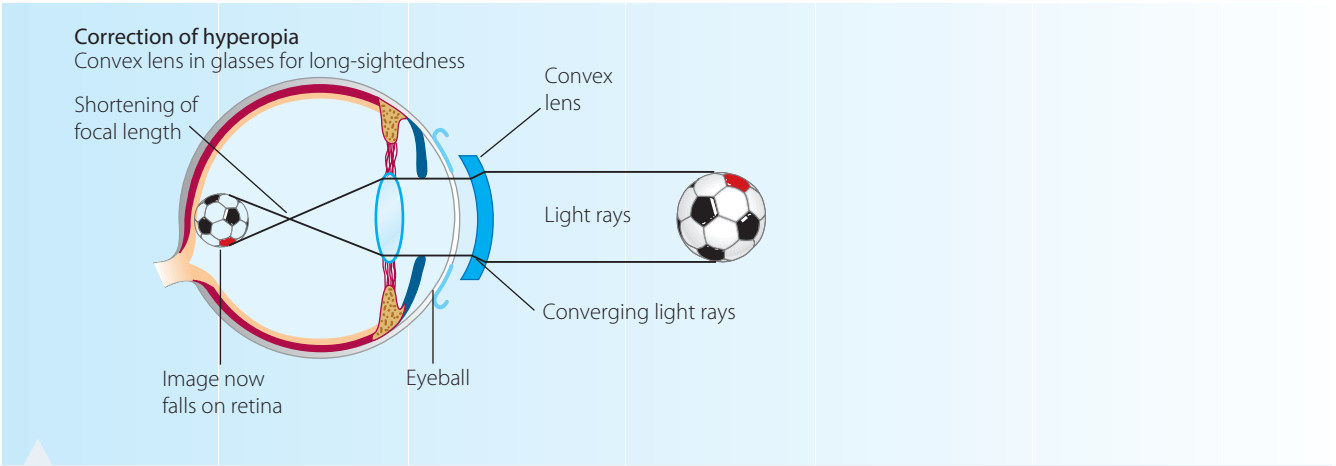


FIGURE 18.17 How corrective lenses work for **a** myopia and **b** hyperopia

Hyperopia can be remedied by wearing spectacles with **convex lenses**, which are thicker towards the centre and thinner towards the edges. This type of lens bends incoming light rays inwards, causing them to begin converging before they reach the eye, shortening the focal length and allowing the focused image of a near object to fall on the retina rather than behind it (Fig. 18.17b).

Contact lenses

Contact lenses are an alternative for those who don't want to wear spectacles all the time and either don't want to undergo or are not suitable candidates for laser surgery. Contact lenses are based on similar technology to spectacles in terms of the shape of the lens and the refraction of light. Although the basic lens structure is convex or concave in shape, it is shaped to fit the curvature of the eyeball. Contact lenses are much smaller than the lenses in spectacles and are worn directly in contact with the surface of the eye.

Cataract surgery

Intraocular lens implantation (IOL) corrects cataracts. The cloudy lens is removed from the lens capsule and an artificial lens is inserted. To do this, the surgeon makes a very small incision in the eye and inserts a device that delivers high-frequency sound that breaks up the cloudy lens. The small lens particles are suctioned out and then an artificial lens is inserted into the lens capsule.

An emerging technology is the use of laser cataract surgery, in which a laser is used to break up the lens and to more accurately place the artificial lens in the lens capsule.

Weblink
Myopia and hyperopia
Watch the video and outline the visual disorders of myopia and hyperopia and how they can be corrected.

Weblink
Cataract surgery
Watch these short videos and prepare a summary of the different types of cataracts and steps involved in cataract surgery.

INVESTIGATION 18.5

A secondary-source investigation of laser surgery in the correction of visual disorders

Information and communication technology capability

Personal and social capability

INTRODUCTION

Many visual disorders of the eye are caused by the inability of the eye to refract the incoming light by the correct amount. Technologies that assist the eye in refracting light to form a focused image on the retina are therefore invaluable. The use of spectacles has already been discussed. Another technology to correct refractive errors of the eye is laser surgery. In this type of surgery, lasers are used to change the curvature of the cornea to enable the light entering the eye to be correctly refracted to form a focused image on the retina. There are different types of laser surgery, the most commonly performed being **LASIK surgery** (laser-assisted in situ keratomileusis).

AIM

To investigate the process of LASIK surgery to assist with the effects of myopia and hyperopia

METHOD

1. Working in pairs or in groups, use a variety of reliable sources to investigate the process of LASIK surgery to correct myopia and hyperopia.
2. Areas to be researched are:
   - suitable candidates
   - reasons why people consider LASIK surgery
   - medical issues that need to be considered
   - the procedures involved in LASIK surgery to correct both myopia and hyperopia
   - side-effects and complications
   - long-term outlook
   - benefits and risks of the procedure
   - other pertinent information.
3. Diagrams or videos of the procedure should be included in your information.
4. Present your information in a manner of your choosing and reference your sources in an acceptable form.
5. Research the advantages and disadvantages of the use of spectacles and contact lenses.
6. Construct a table to summarise the advantages and disadvantages of the use of contact lenses, spectacles and laser surgery to correct refractive errors in the eye.

RESULTS

1. Presentation of LASIK surgery as a means of assisting with visual disorders caused by refractive errors in the eye.
2. Presentation of a table summarising the advantages and disadvantages of the use of contact lenses, spectacles and laser surgery to correct refractive errors in the eye.
3. Reference your sources in an acceptable form.

DISCUSSION

1. Describe the impact of this procedure on the affected individual.
2. Discuss the advantages and disadvantages of laser surgery compared with wearing contact lenses or spectacles.
3. Assess the relevance, reliability, validity and accuracy of the information you have collected.
4. Identify ways in which this investigation could have been improved.

CONCLUSION

Write summary sentences to address the aim of this investigation.

The bionic eye

Bionic Vision Technologies is developing a bionic eye to restore vision to people with retinitis pigmentosa (a disease where the rods and cone cells degenerate gradually) and age-related macular degeneration. The bionic eye consists of a camera attached to a pair of glasses, which transmits high-frequency radio signals to a microchip implanted in the eye (Fig. 18.18). Electrodes on the implanted chip convert these signals into electrical impulses to stimulate cells in the retina to convert light rays into electrical impulses, which are then sent to the brain via the optic nerve.

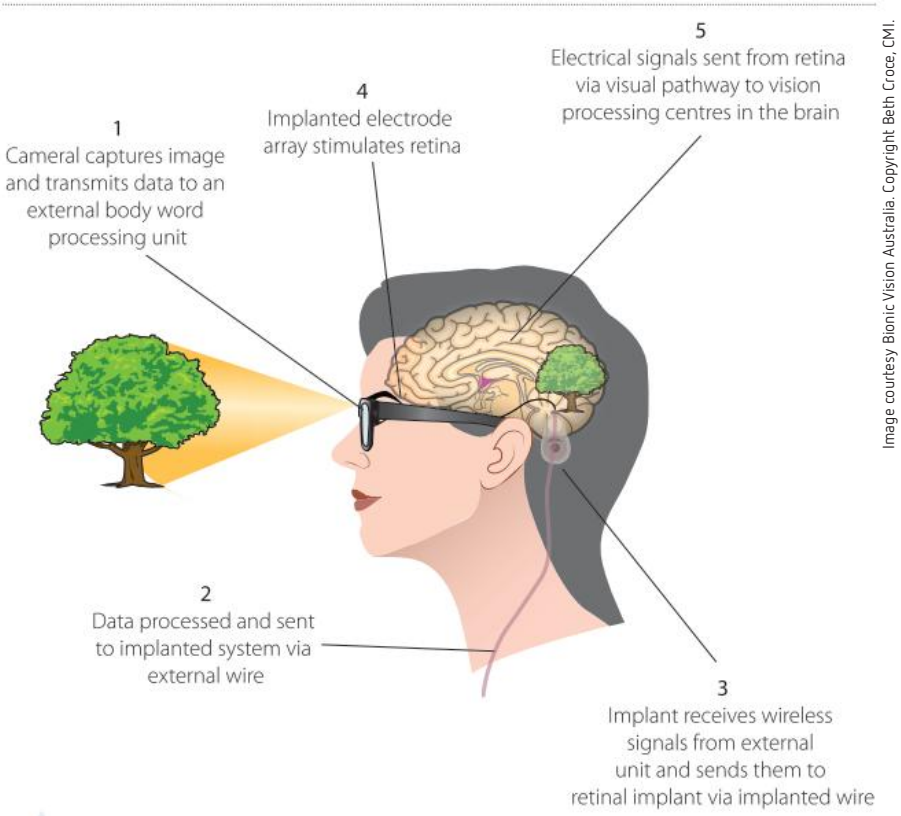


FIGURE 18.18 How the bionic eye works

KEY CONCEPTS

- Lenses are used in spectacles and contact lenses to correct the refractive errors of the eye.
- Concave lenses correct myopia, while convex lenses correct hyperopia.
- Laser surgery changes the convexity of the cornea to correct refractive errors and allow light rays to be focused on the retina.
- Cataract and laser cataract surgery restore vision to cataract sufferers.
- The bionic eye could help to restore vision to those suffering from retinitis pigmentosa by bypassing damaged rod and cone cells.

CHECK YOUR UNDERSTANDING 18.2c

1 Explain, with the use of a diagram, how spectacles can be used to correct:
   a myopia
   b hyperopia.
2 Describe another technology besides the use of lenses that can be used to correct refractive errors of the eye.
3 Compare the processes of cataract surgery and laser cataract surgery.
4 Compare cochlear implants with the bionic eye.

18.3 The kidney

The **kidneys** are organs that form part of the excretory system. Their function is to remove nitrogenous wastes from the bloodstream and maintain optimal levels of substances such as salt and water (**osmoregulation**).

Structure and function of the kidney

The kidney has three main functions: **filtration**, **reabsorption** and **secretion**. To carry out these functions, the kidney has three main regions: the cortex, medulla and pelvis.

Weblink
Structure and function of the kidney

The functional unit of the kidney is the **nephron** (Fig. 18.19). Each kidney contains millions of these microscopic units, extending from the cortex of the kidney and into the medulla. Each nephron consists of a **Bowman's capsule**, which is a spherical, two-sided, hollow, cuplike structure that contains a spherical network of capillaries, called the **glomerulus**. The far side of the Bowman's capsule leads into the tubules that make up the rest of the nephron. These tubules are the **proximal tubule**, the **loop of Henle** and the **distal tubule**, which leads into the **collecting duct**.

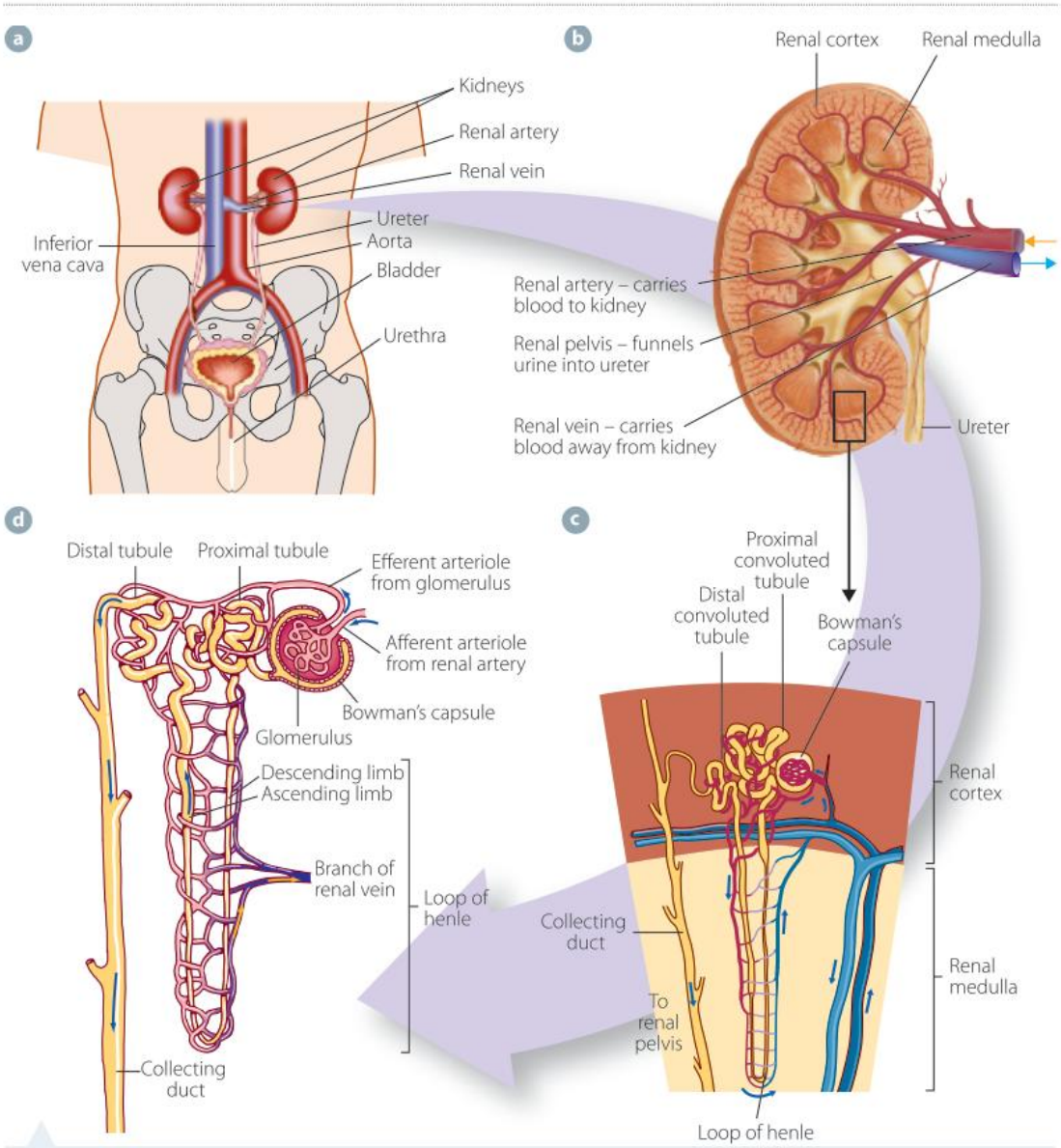


FIGURE 18.19 **a** Excretory system of mammals; **b** macroscopic structure of mammalian kidney (longitudinal section); **c** microscopic structure showing the distribution of tubules in the mammalian kidney; **d** nephron and associated capillaries

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Filtration

Under high pressure, blood travels from the heart via the aorta to the renal artery and into the kidney. The renal artery branches into smaller and smaller vessels until millions of capillaries are formed. Each capillary enters a nephron, where it forms the glomerulus inside the hollow of each Bowman's capsule. Filtration is based on the size of substances, so all substances in the glomerular blood that are small enough will pass through the walls of the glomerulus into the Bowman's capsule. This includes nitrogenous waste products and large volumes of water carrying dissolved substances including amino acids, glucose and salts (ions).

Once inside the Bowman's capsule, this fluid, now known as the **glomerular filtrate**, continues its journey along the tubules that extend at the back of the capsule (Fig. 18.20). Larger molecules, such as proteins and blood cells, remain in the glomerular blood.

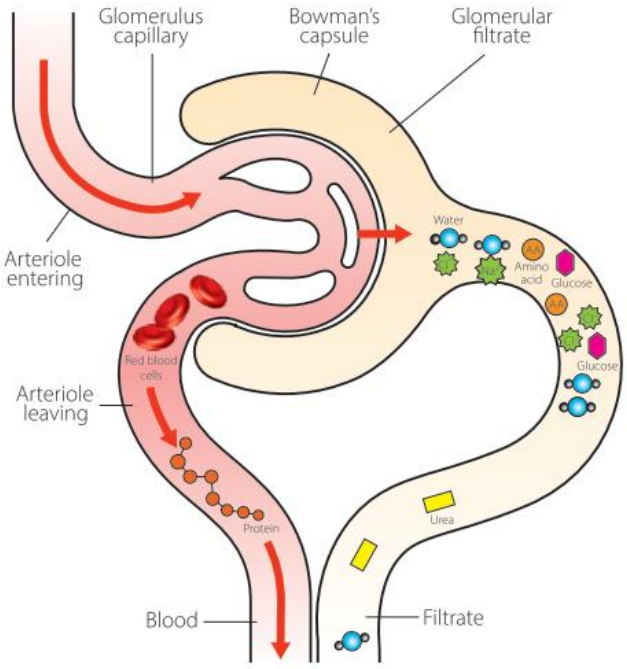


FIGURE 18.20 Filtration of blood in the Bowman's capsule

Reabsorption

Reabsorption returns essential components that have been filtered out of the blood back into the bloodstream. Amino acids, glucose, varying quantities of ions (such as sodium (Na^+), potassium (K^+), chloride (Cl^-), calcium (Ca^+) and hydrogen bicarbonate (HCO_3^-) and some vitamins are reabsorbed into the bloodstream (Fig. 18.21). The differing rates of reabsorption of particular ions depend on feedback from the body. All solutes that are reabsorbed from the nephron move by *active transport* and *facilitated diffusion* in both the proximal and the distal tubule (Fig. 18.22).

Glomerular filtrate also contains a relatively high concentration of dissolved urea and other wastes, most of which are not reabsorbed.

As the solutes are reabsorbed, water follows by the passive process of *osmosis*. In the ascending loop of Henle and the collecting duct, a large number of ions (mostly sodium) are actively pumped into the tissues of the medulla. This causes the concentration of water in these tissues to be much lower than in the tubules. As a result, water moves from the descending loop of Henle and the collecting duct into the tissues by the process of osmosis. The reabsorption of water occurs in all parts of the tubules and collecting duct, except the ascending loop of Henle.

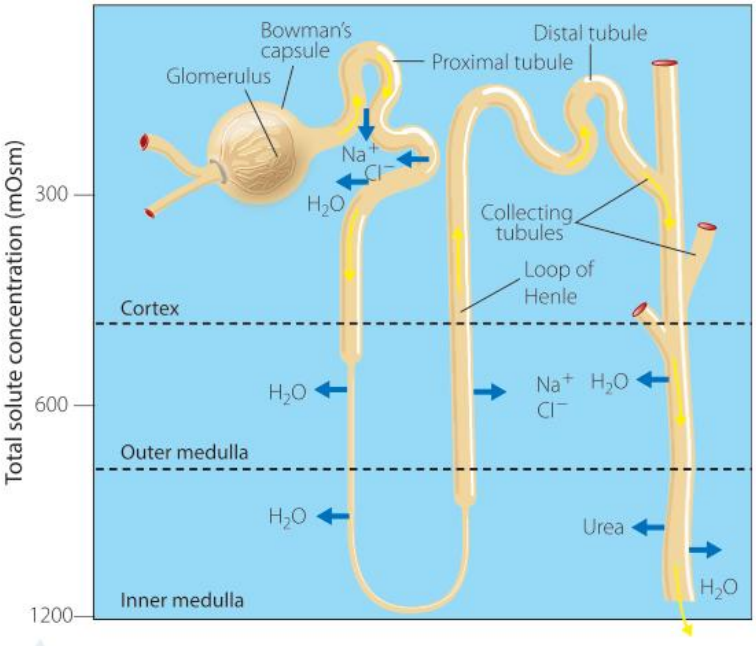


FIGURE 18.21 Reabsorption of water and various ions along the tubule. Blood vessels are not shown.

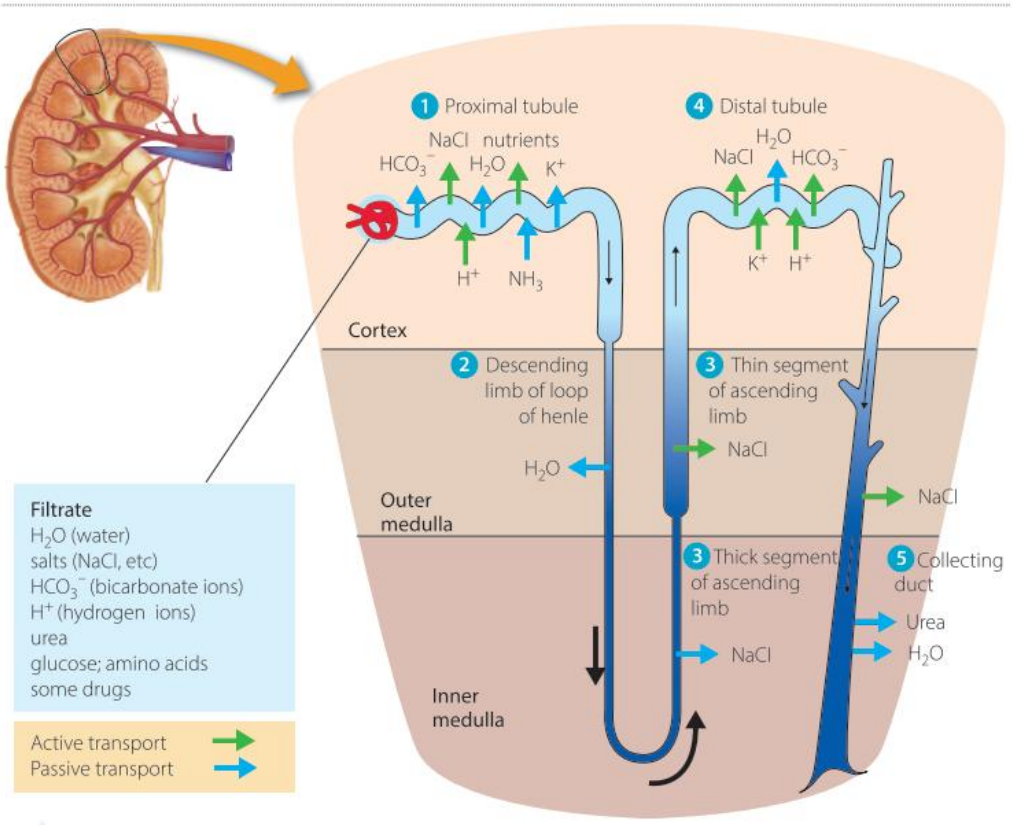


FIGURE 18.22 Reabsorption of ions and water from the tubules by active transport (ions) and passive transport (water). Blood vessels are not shown.

Secretion

Secretion (or tubular secretion) is the third process that contributes to urine formation in the nephron. Secretion involves the removal of toxic substances from the blood capillaries and tissues surrounding the tubules and their active movement into the tubules for removal. This includes metabolic wastes such as urea, uric acid, ammonia and hydrogen ions, along with drugs such as penicillin, saccharin and morphine. Movement of urea and ammonia is mainly by means of diffusion, whereas all other secretion involves active transport.

Hydrogen ions (H^+), saccharin, and *drugs* such as penicillin and morphine, are secreted into the proximal part of the nephron. *Urea* is secreted into the descending limb of the loop of Henle (the ascending limb and distal tubules are impermeable to urea).

Hormonal regulation of osmoregulation in the kidney

The hormone aldosterone stimulates the reabsorption of salt in the loop of Henle, to regulate salt and water balance in the kidney.

The hormone antidiuretic hormone (ADH or vasopressin) stimulates the reabsorption of water in the kidney.

Hormonal control of osmoregulation in the kidney is dealt with in more detail in Chapter 14 (pages 490–1).

Removal

The water, nitrogenous wastes and other wastes that remain in the collecting duct are known as urine. Urine collects in the pelvis of the kidney before moving through the ureters to the bladder, after which it is eliminated from the body through the urethra.

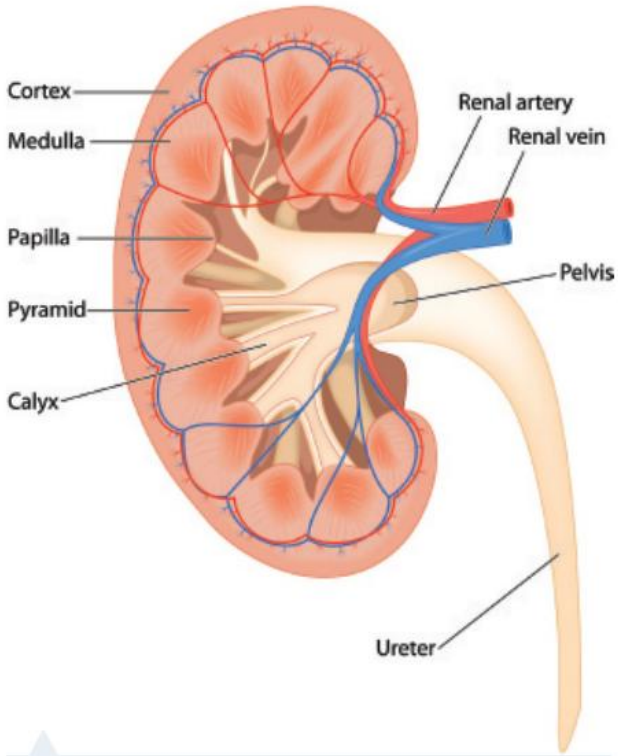
INVESTIGATION 18.6

A first-hand investigation to identify the regions of the mammalian kidney

INTRODUCTION

This investigation may be undertaken as a dissection, using fresh kidneys, or as an interactive virtual dissection. Models of the kidney, both physical and digital, can be used as well.

The kidneys lie on either side of the midline on the back wall of the abdomen, in the region of the waist. In humans, each kidney is bean-shaped, about 10.5–13 cm long, 6 cm wide and 3 cm thick. Regions to be identified are shown in Figure 18.23.



Blamb/Shutterstock.com

FIGURE 18.23 Longitudinal section of the kidney, indicating the different regions

AIM

To identify the regions of the mammalian kidney involved in the excretion of wastes, and relate these structures to their functions

MATERIALS

- dissecting tray
- newspaper
- scalpel
- forceps
- probe
- scissors
- kidney
- gloves
- models of kidney
- pins and label flags

RISK ASSESSMENT

Draw up a table that identifies three hazards associated with this investigation, the risk these hazards pose and how these risks can be safely managed.

METHOD

1 Work in pairs or in groups of four. Lie the kidney on the dissecting tray.

External structure

2 Examine the external structure of the kidney, noting its surrounding fat (**adipose tissue**). Remove the fat, leaving the vessels at the **hilum** intact. The hilum is the curved indentation of the kidney where the blood vessels enter and leave the kidney and the ureter leaves the kidney.

3 Compare the size of the kidney that you have for dissection with the dimensions given for an average human kidney.

4 Identify the vessels, distinguishing between the **renal artery**, **renal vein** and ureter if present.

5 Detach the **renal capsule**. Describe its appearance and function.

6 Draw a life-sized diagram of the external structure of the kidney.

Internal structure

7 Cut the kidney in longitudinal section, making an incision along the side opposite the hilum. Note the opening to the ureter. Insert a probe through the hole and observe where it exits.

»

8 Identify the regions of the kidney: outer **cortex**, **medulla** and **renal pelvis**. Compare the colour and appearance of the cortex and the medulla.

9 Insert a probe below the **renal pyramids**, slip the lower blade of the scissors into the gap and slit through each pyramid to follow the path of the **calyces**. Urine from the collecting tubules drains into these calyces, which carry the urine to the renal pelvis, ureters and **bladder**.

10 Draw a diagram of a longitudinal section through the kidney, showing the internal structure as seen in the dissection. Annotate the diagram by writing the function of each structure labelled (a minimum of six structures). Do *not* draw the textbook diagram.

11 Identify each of the following regions of the kidney, using toothpicks with coloured flags:

- red flag – the part of the kidney containing collecting tubules
- blue flag – the region of the kidney containing glomeruli and Bowman's capsules
- yellow flag – the part of the kidney that carries urine from the collecting tubules.

DISCUSSION

1 In a table like the one below, list the parts of a nephron that you would expect to find in the cortex and the parts you would expect to find in the medulla:

glomerulus, Bowman's capsule, first convoluted tubule, loop of Henle, second convoluted tubule, collecting tubule, blood capillaries

REGION OF KIDNEY	PARTS OF NEPHRON
Cortex	
Medulla	

2 Give the correct biological term for each of the following two functions of the kidney:

a filter out wastes such as urea

b regulate the water and salt balance.

3 What adjective is used to describe anything associated with the kidney?

4 Compare the chemical composition of blood in the renal artery with that of the renal vein.

CONCLUSION

Write a few summary sentences to address the aim of this investigation.

KEY CONCEPTS

- The main functions of the kidney are to excrete nitrogenous wastes and to balance the level of water and salt in the blood.
- The kidney contains three main regions: the cortex, medulla and pelvis.
- The nephrons are the functional units of the kidney. Each kidney contains millions of nephrons.
- Nephrons contain the glomerulus, Bowman's capsule, proximal tubule, loop of Henle, distal tubule and collecting duct.
- Filtration occurs in the junction between the glomerulus and the Bowman's capsule. All substances that are small enough move into the tubule from the bloodstream.
- Reabsorption of all substances required by the body occurs along the tubules and in the collecting duct.
- Secretion of additional wastes into the tubules occurs along the length of the nephron.
- Remaining wastes and water collect in the pelvis of the kidney to form urine and move via the ureters to the bladder before elimination from the body via the urethra.
- Water and salt conservation in the kidney are stimulated by ADH and aldosterone respectively.

CHECK YOUR UNDERSTANDING 18.3

1 Identify:
   a the main functions of the kidney
   b the main functional unit of the kidney.
2 Draw a diagram of:
   a a cross-section of a kidney, with the following regions labelled: cortex, medulla, pelvis
   b a nephron, with the following labelled: glomerulus, Bowman's capsule, proximal tubule, loop of Henle, distal tubule, collecting duct.
3 On the diagram of the nephron, identify and label the area where the following occurs:
   a filtration
   b reabsorption of essential components
   c secretion of toxins
   d active pumping of salts into the tissues followed by movement of water by osmosis.

Loss of kidney function

Loss of kidney function often occurs gradually, with minimal signs until kidney function is significantly impaired. The causes are many and often involve damage to the individual filtering units, the nephrons. If the nephrons are damaged they cannot carry out their filtering, reabsorption and secretion functions properly. This leads to a failure to remove wastes effectively and an inability to balance water and salt levels in the blood.

Symptoms of loss of kidney function include:

- nausea
- vomiting
- loss of appetite
- fatigue and weakness
- sleep problems
- changes in the volume of urine produced
- decreased mental sharpness
- muscle twitches and cramps
- swelling of feet and ankles
- persistent itching
- chest pain, if fluid builds up around the lining of the heart
- shortness of breath, if fluid builds up in the lungs
- high blood pressure (hypertension) that is difficult to control.

Some of the causes of kidney disease leading to reduced function are:

- diabetes type 1 and 2 – high levels of blood glucose cause the kidneys to filter more blood, which causes stress on the nephrons. The nephrons eventually become damaged and larger molecules, such as proteins, 'leak' into the tubules and cause blockages and further damage to the nephrons
- continued high blood pressure – the nephrons are damaged by the constant high force of the blood being pushed through the walls of the blood vessels of the glomerulus into the Bowman's capsule. This causes permanent damage to the nephrons, again with large molecules 'leaking out' into the tubules, causing blockages and further damage, which reduces the ability of the nephrons to function effectively
- recurrent kidney infections – this causes damage to the nephrons, and this damage as well as the prolonged use of certain medications and drugs to treat the infections can reduce kidney function.

- kidney stones, tumours or anything that blocks the passage of urine – causes the urine to 'back up' into the kidney, which causes a build-up of pressure that, unless the blockage is removed, will cause kidney damage leading to a complete shutdown.

Technology to assist with loss of kidney function

The kidneys are responsible for filtering our blood and removing metabolic wastes so that they are excreted in the urine. The kidneys are also responsible for maintaining the balance of water and salts in the blood. If a person suffers from kidney failure, there is no natural means by which these wastes can be removed and their toxic effect will eventually lead to death.

The process of **renal dialysis** has been developed to carry out some of the functions of kidneys, so that blood may be effectively filtered even when the kidneys are damaged (Fig. 18.24). There are two types of renal dialysis: **haemodialysis** and **peritoneal** dialysis (Fig. 18.25).

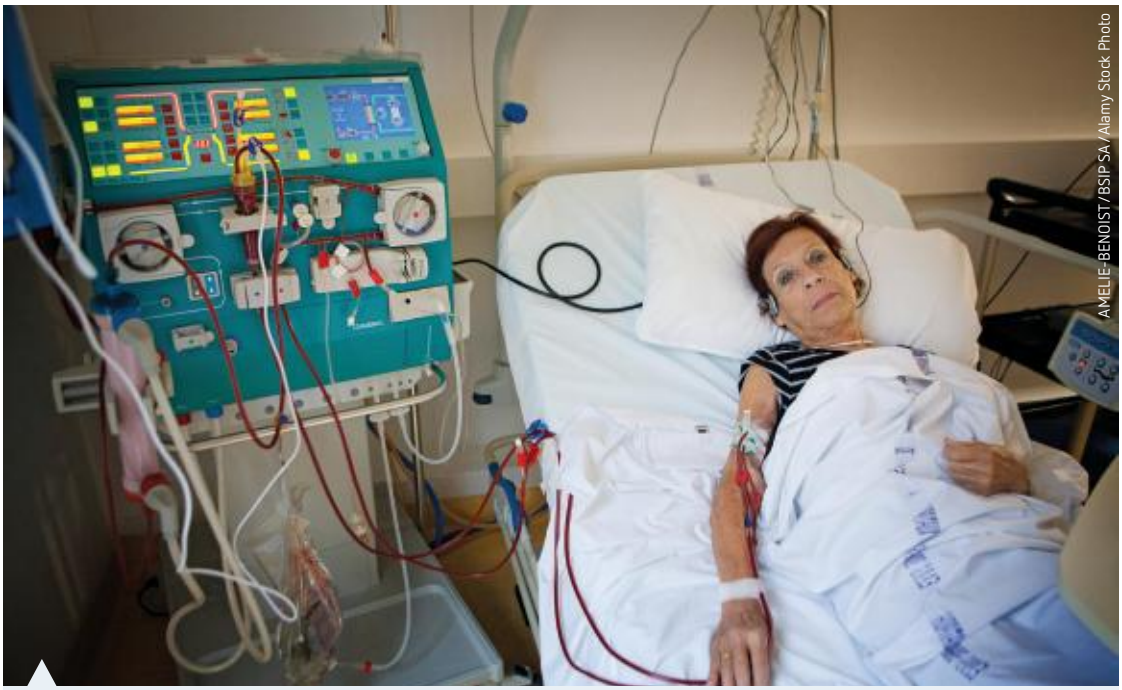


FIGURE 18.24 A patient undergoing renal dialysis

Weblink
Dialysis
Draw a flow chart of the process of haemodialysis

The main function of a dialysis machine is to remove metabolic wastes that have built up in the person's blood. The patient is connected to a dialysis machine, which pumps their blood through a system of tubes (coiled to increase their surface area and therefore the rate of diffusion), which have artificial semi-permeable membranes. The tubes are submerged in dialysis fluid (**dialysate**), which flows in the opposite direction to the blood in order to maintain a concentration gradient to maximise diffusion. Dialysate has the same concentration as blood plasma, without the metabolic wastes. Because the concentration of metabolic waste is higher in the blood than in the dialysing fluid, the waste materials move through the semi-permeable membrane into the dialysing fluid by diffusion. Continual replacement of the dialysate is required.

Renal dialysis must be done for 3–4 hours, two or three times per week. As well as its time-consuming nature, another disadvantage of the process is that only limited amounts of fluid/wastes can be removed from the blood; other substances such as sodium phosphate and potassium ions do not diffuse quickly enough and therefore may accumulate in the blood. It is therefore recommended that patients follow a specific diet to prevent this, as renal dialysis is not effective in regulating the concentration of these ions in the blood.

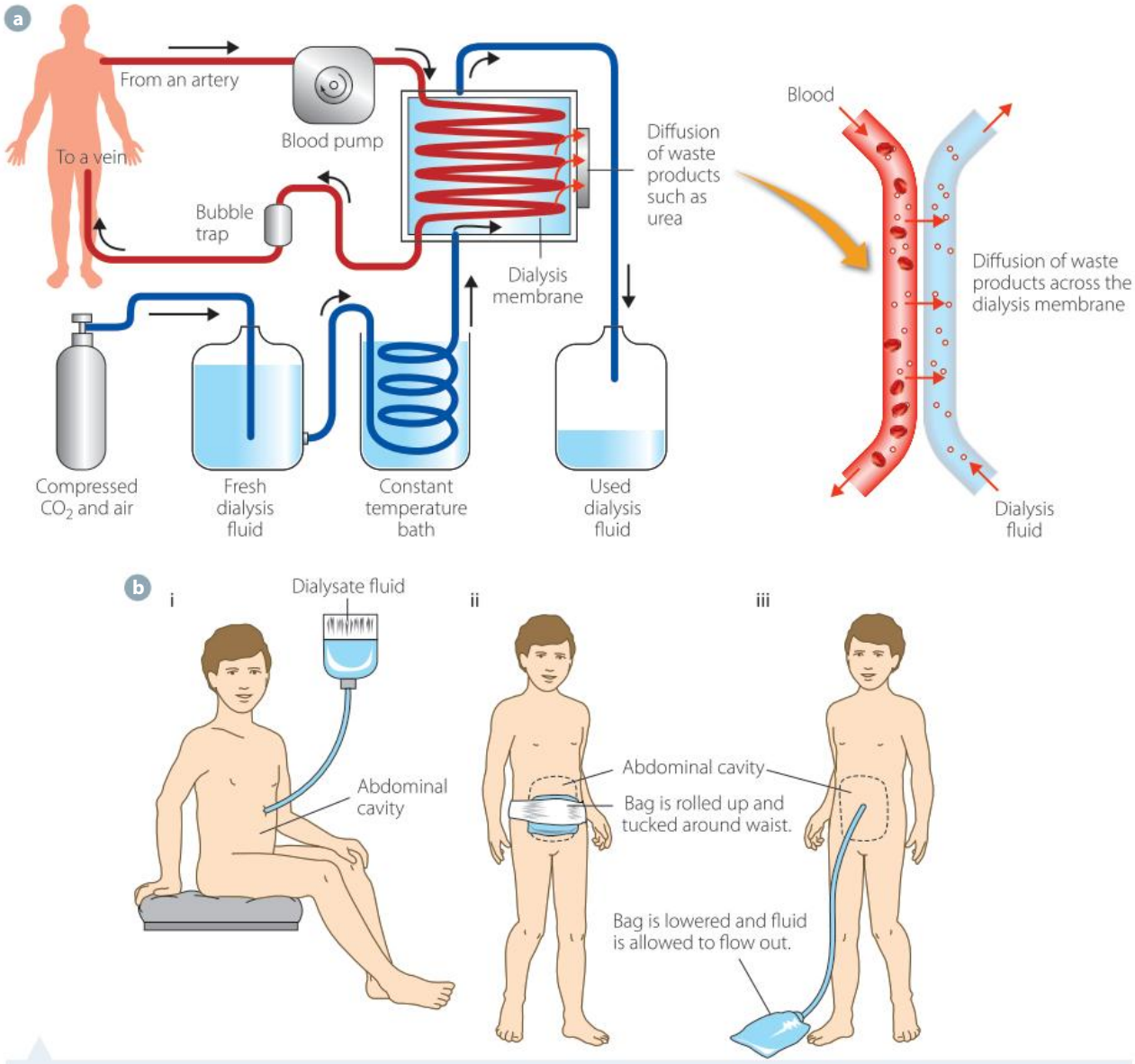


FIGURE 18.25 Renal dialysis: **a** haemodialysis; **b** peritoneal dialysis

INVESTIGATION 18.7

A secondary-source and first-hand investigation to research renal dialysis

INTRODUCTION

Dialysis is a technology that can assist the body to remove wastes when a person experiences a loss of kidney function. In this investigation you will describe the two types of dialysis and state when each may be used. You will also discuss the advantages and disadvantages of dialysis and assess whether it is a long-term replacement for normal kidney functioning. If possible, a visit to a renal dialysis unit will contribute to understanding the issues associated with use of dialysis.

Personal and social capability

» Your group should plan this investigation, deciding on the aim, the type of information to be researched to address the points above, and how to present the information. Questions to be addressed in the discussion should be composed and then answered.

If a visit to a renal dialysis unit can be organised, then questions to be posed to dialysis patients and nursing staff that are sensitive to their situation should be prepared beforehand.

RESULTS

Present your results in a format agreed to with your teacher.

18.4 Effectiveness of technologies

Technologies aim to assist with the effects of a disorder and improve the quality of life of the sufferer. Without technology, individuals would have to manage to the best of their ability to cope with the effects of the disorder. Not only would these individuals be affected, but so too would their families or carers. The burden on the health and welfare systems would be increased due to the continual support that would have to be provided.

The effectiveness of the cochlear implant for those with sensorineural hearing loss is an example of a highly effective technology. The cochlear implant gives individuals the ability to communicate effectively with those around them, rather than, as previously happened, having to find other ways of communicating. These involved the use of sign language, lip reading and often a reduced ability to develop effective language skills, especially in those born profoundly deaf. This led to a reduced quality of life and a limited ability to function at higher levels in the community.

With the advent of the cochlear implant, those born profoundly deaf now have the ability to develop at a normal rate and show no signs of developmental delays in language, hearing and speech. Infants as young as six months can have cochlear implants fitted and, with support and guidance, will develop language and speaking skills normally. There is no need to attend special schools and they can be fully integrated into society.

Older adults who have lost most of their residual hearing and have a cochlear implant are able, after a period of adjustment and training, to hear the world around them again and experience an improved quality of life. They are able to be more independent and to contribute positively to both their family and society without extra assistance.


Sputnik/Science Photo Library

FIGURE 18.26 A cochlear ear implant enables people to interact fully with their environment.

The disadvantages associated with the use of cochlear implants include:

- having to undergo an operation for the implantation of the device
- post-operative side effects
- ongoing costs for updating
- programming and training required to teach the person to interpret the sounds they hear
- a limited distance of hearing.

These disadvantages are far outweighed by the advantage of being able to hear, develop speech normally and be a highly functioning and contributing member of society with no real disability.

INVESTIGATION 18.8

A secondary-source investigation to evaluate the effectiveness of a technology

INTRODUCTION

Many technologies that assist with the effects of a disorder have been studied. You will be required to judge the effectiveness of one of these technologies based on a set of criteria.

Ethical understanding

Literacy

1. Working in pairs or groups, develop an aim for this investigation.
2. Set out a method to be followed. To do this, decide what criteria you will use to make your judgement about the effectiveness of your chosen technology.

   Things such as the advantages and disadvantages of the use of the technology, along with what is involved with the use of the technology and the effects on the individual if the technology is not used, are areas that can be investigated.
3. Your results will contain the information you have gathered. Decide on an appropriate format for your results.
4. The discussion will provide a judgement and the evidence that supports your judgement of the effectiveness of the technology.
5. A conclusion will finalise your investigation.

KEY CONCEPTS

- Loss of kidney function can occur for numerous reasons, including diabetes, high blood pressure, kidney infections and blockages.
- Most of these conditions cause damage to the fragile nephrons.
- Dialysis uses an 'artificial kidney' to remove urea from the blood when kidney function is insufficient to do so.
- Dialysis can also balance the water and salt levels to some extent.
- There are two types of dialysis: haemodialysis and peritoneal dialysis.
- Renal dialysis patients undergo dialysis several times a week for extended periods.
- The effectiveness of a technology can be assessed on the basis of certain criteria.

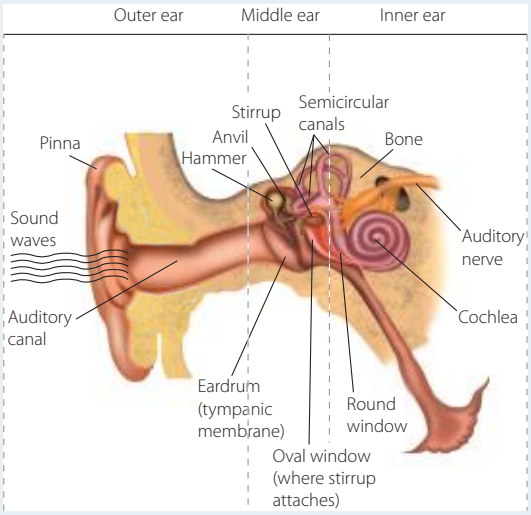
CHECK YOUR UNDERSTANDING 18.4

1. Explain how different causes of loss of kidney function affect the nephrons.
2. Outline the concentration of substances in dialysate fluid compared to the blood of the patient, and explain the significance of this concentration.
3. Outline the conditions under which dialysis is used and describe how it removes urea from the blood.
4. Describe the effects of dialysis on the lives of those who suffer from reduced kidney function.
5. List three criteria that could be used to assess whether a technology designed to assist with a disorder is effective.

18 CHAPTER SUMMARY

Technologies and disorders: How can technologies be used to assist people who experience disorders?

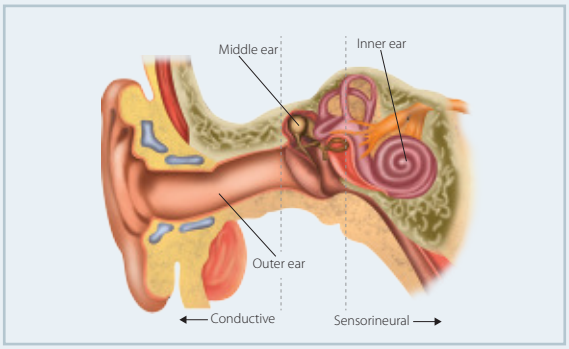
THE EAR



Pathway of sound

External auditory canal
↓
Tympanic membrane
↓
Ossicles
↓
Oval window
↓
Cochlea
↓
Round window

Hearing loss

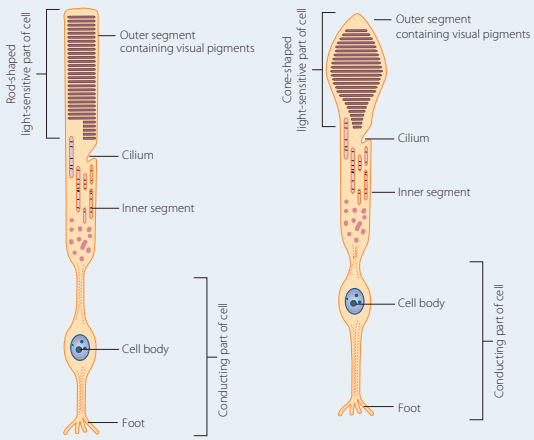


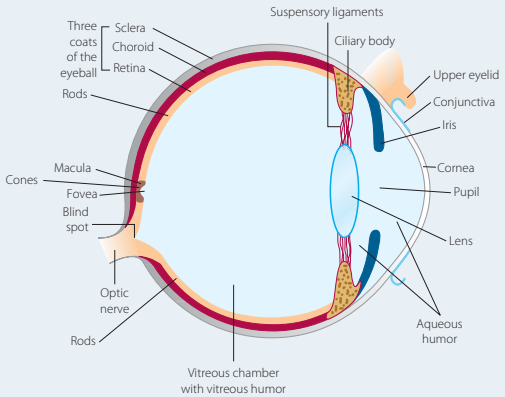
Technology to assist those with hearing loss

- Hearing aid (used for both conductive and sensorineural hearing loss)
- Bone conduction implant (conductive hearing loss)
- Cochlear implant (sensorineural hearing loss)

THE EYE

Photoreceptor cells: rods and cones
Convert light into electrochemical impulses
Rods – rhodopsin pigments – detect light
Cones – iodopsins – colour vision





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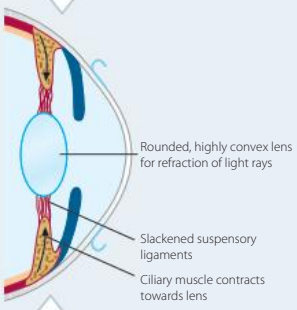
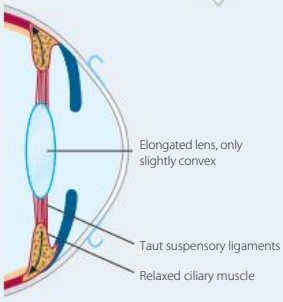
Accommodation

Process by which the lens changes its curvature to focus on objects at different distances

Near vision



Distant vision



Visual disorders

- Hyperopia – can focus on distant objects, not close
- Myopia – can focus on close objects, not distant
- Cataracts – cloudy lens
- Macular degeneration – degeneration of cells beneath the retina

Technologies to assist those with visual disorders

Spectacles, contact lenses concave lenses for myopia, convex lenses for hyperopia

LASIK surgery changes curvature of the cornea to correct refractive errors of eye

Cataract surgery (intraocular lens implantation) replaces cloudy lens with artificial lens

THE KIDNEY

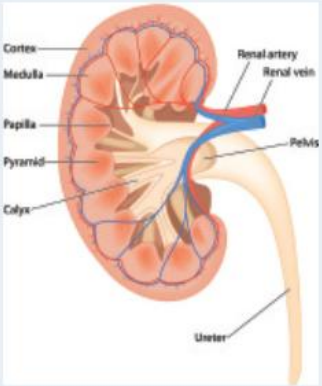
The kidney removes nitrogenous waste from the bloodstream and carries out osmoregulation by filtration, reabsorption and secretion.

Functions

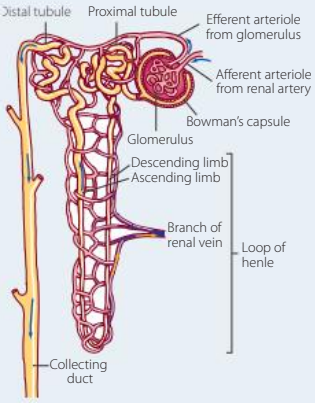
- Filtration
- Reabsorption
- Secretion

Wastes and water collect in pelvis of kidney to form urine.

Three main regions – cortex, medulla, pelvis



Nephron – functional unit of kidney (millions in kidney)



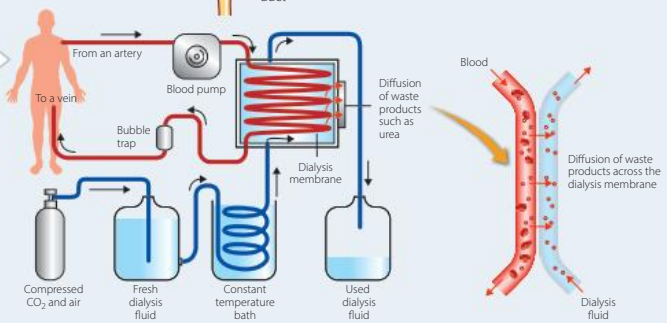
Technology to assist with kidney disorder

Loss of kidney function

Leads to failure to remove wastes and inability to balance water and salt levels in blood.

Technology to assist those with loss of kidney function

Renal dialysis – carries out some of the functions of the kidney



18 CHAPTER REVIEW QUESTIONS

Review quiz

1 A newborn baby is not responding normally to the sounds around him. On testing, it is found that he is profoundly deaf. The cause of this deafness is genetic and affects the hair cells in his cochlea. It is also found that his auditory nerve is functioning correctly.

Outline the technology that can be used to assist with this type of hearing disorder. In your answer, include the advantages of early intervention.

2 A football player received a knock to the back of his head and consequently had vision problems for a number of days. Explain why the football player developed visual disturbance.

3 **a** Explain, with the use of diagrams, why people tend to hold their reading material further and further away from their eyes as they get older.

b What is the name given to this disorder?

c Describe one technology that could be used to assist with this disorder.

4 **a** Describe what is meant by 'refraction of light'.

b Outline the structures in the eye through which light is refracted.

c Why is it important that light is refracted in the eye?

5 Write a paragraph to compare the accommodation of the eye for near vision and for distant vision. Include in your description a comparison of the refractive power of the lens, from rest to maximum accommodation.

6 Cataract surgery replaces the old clouded lens with a clear lens that allows the light to pass through unhindered. Critically evaluate the consequences of cataract surgery on the quality of life of an elderly person.

7 Justify the directive, given by the supervisor on a building site, that all workers must wear earplugs when machinery is being used.

8 Outline the special dietary requirements of patients undergoing dialysis, and explain why it is necessary to follow these strict dietary guidelines.

9 Evaluate the impact of undergoing dialysis on the lifestyle of a patient who has lost kidney function.

10 Explain why candidates for the 'bionic eye' are tested for a functioning optic nerve.

11 Compare the functioning of the kidney with the process of dialysis.

12 Explain how refractive errors in the eye cause visual disorders.

13 Prepare an answer to the inquiry question: 'How can technologies be used to assist people who experience disorders?'

14 Compare the functioning of a cochlear implant with that of a bionic eye.

Exam preparation

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MODULE 8: NON-INFECTIOUS DISEASE AND DISORDERS

Answer the following questions.

1 **a** Define 'homeostasis'.

b Describe how homeostasis maintains the internal environment, using humans as an example. Refer to negative feedback loops and examples such as the regulation of temperature and blood glucose levels to assist your description.

2 Outline the adaptations that you would expect to find in an endothermic organism that lives in a hot, dry environment. Justify the adaptations you have identified.

3 A plant was found to have a covering of light hairs on its small leaves, a lighter colour on the underneath surface of the leaves and sunken stomata. Predict the environment in which this plant would be found, and explain how these adaptations assist it to survive successfully in this environment.

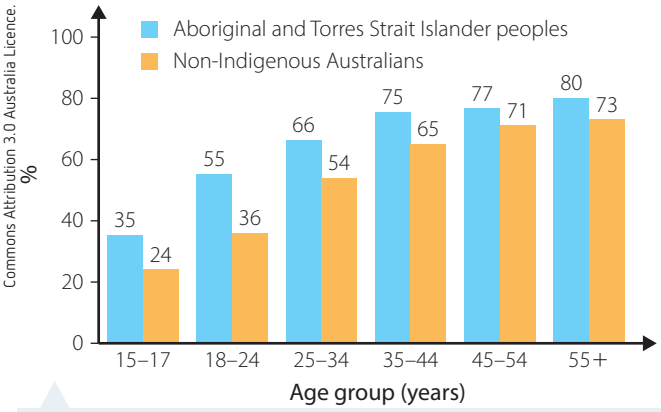
4 **a** Draw a labelled diagram of a motor neuron and indicate the direction of movement of a nerve impulse.

b Construct a table to distinguish between the structure and function of the following types of neurons: sensory, motor, interneuron.

5 How do the nervous and endocrine systems work together in maintaining an organism's internal environment? Use specific examples to illustrate your answer.

6 Construct a table to summarise the cause and effects on the body of one non-infectious disease, from each of the following types: genetic disease, nutritional disease, cancer, diseases caused by environmental exposure.

7 The graph below shows the proportion of people in Australia, aged 15 years and over, who were overweight or obese, by Indigenous status and age, 2012–13.



Proportion of people in Australia,15 years and over, who were overweight or obese, by Indigenous status and age, 2012–13

a Use the graph to compare the prevalence of obesity in non-Indigenous Australians to Aboriginal and Torres Strait Islander peoples in the respective age groups, for 2012–2013.

b Suggest reasons for the differences outlined in part **a**.

8 For an identified non-infectious disease caused by exposure to the environment, or a nutritional disease, outline:

a the treatment/management

b future directions for further research.

9 **a** Outline an example of an epidemiological study.

b Comment on the reliability and validity of this study.

10 Evaluate, using examples, the benefits of conducting an epidemiological study.

11 Outline one educational campaign designed to prevent a non-infectious disease and evaluate its effectiveness.

12 A poster was produced to educate primary school-aged children about the importance of a healthy diet and exercise, to prevent them becoming overweight or obese. What would the poster designers need to take into account when deciding how to design the poster and what material to include in the poster?

13 **a** Outline one example of how genetic engineering can be used to prevent a non-infectious disease.

b Discuss any ethical issues associated with the use of the genetic engineering technique outlined in **a**.

14 **a** Relate the structure and functioning of the eye to the causes of the visual disorders of myopia and hyperopia. Use diagrams to aid your explanation.

b Describe how different technologies can be used to assist with these disorders.

c Evaluate the effectiveness of these technologies against three chosen criteria.

15 Describe how cochlear implants can assist with sensorineural hearing loss, and relate their functioning to the normal functioning of the ear.

16 One of your acquaintances has a serious kidney disease that has affected the nephrons and has to undergo haemodialysis.
Create a pamphlet explaining how haemodialysis could assist with the effects of this disease and how it differs from the normal functioning of the kidney.

17 Explain the role of hormones in regulating kidney functioning.

DEPTH STUDY SUGGESTIONS

- Investigate the use of gene therapy in the treatment of non-infectious disease and the associated ethical and scientific debates.
- Design and construct a model, different from the one used in Investigation 18.4, to demonstrate the process of accommodation in the mammalian eye.
- Research the use of cell therapy to genetically modify a patient's immune cells to recognise and kill cancer cells.
- Create a presentation that explains the reasons that cancer is so hard to study and cure.
- Discuss the use of cell lines in scientific research and the importance of correct identification of these lines in ensuring the validity of scientific studies.
- Investigate the development and use of the different types of 'bionic eye' used to restore sight to individuals who have visual disorders.
- Research the creation and uses of blood stem cells to treat and cure diseases.
- Compare the changes in the leading causes of death over the past 20 years in both developed and developing regions of the world. Present your information in digital form, which could include interactive graphs and maps.
- Design and produce a large classroom poster to summarise the adaptations exhibited by ectotherms to allow them to function in an environment with widely fluctuating temperatures.
- Investigate a non-infectious disease that interests you, including the cause, symptoms, treatment, occurrence in different regions of the world and any possible preventative measures. Create an interactive web page or other form of digital media to present the information about this disease.

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ANSWERS

TRY THESE YOURSELF

WORKED EXAMPLE 5.1

1 a all Bb; all black

b 1 BB : 2 Bb : 1 bb; 3 black, 1 white

c 1 Bb : 1 bb; ½ black, ½ white

2 a Bb

b

	B	b
B	BB	Bb
b	Bb	bb

c 2/3 of the curly-winged flies would be heterozygous

WORKED EXAMPLE 5.2

1 b Skippy's dad – Aa, Skippy's mum – aa, Bouncer's dad – AA or Aa, Bouncer's mum – AA or Aa
Skippy – Aa, Bouncer – Aa, Joey 1 – aa, Joey 2 – Aa or AA

WORKED EXAMPLE 5.3

1

	GENERATION 1	GENERATION 2	GENERATION 3
Genotype frequency of: bb BB	 5/6 = 0.83 1/6 = 0.17	 3/6 = 0.50 3/6 = 0.50	 1/6 = 0.17 5/6 = 0.83
Phenotype frequency of: White moths Black moths	 5/6 = 0.83 1/6 = 0.17	 3/6 = 0.50 3/6 = 0.50	 1/6 = 0.17 5/6 = 0.83
Allele frequency of: B b	 2/12 = 0.17 10/12 = 0.83	 6/12 = 0.5 6/12 = 0.5	 10/12 = 0.83 2/12 = 0.17
Possible reasons for change	White moths are more easily seen by predators and are more likely to be eaten; the black moth has a selective advantage.		
Hypothesis	If a moth is black, then its rate of capture by predators will be less frequent.		

2

FREQUENCY TYPE	CALCULATIONS
Genotype frequency: AO BO AB OO	 2/5 = 0.40 1/5 = 0.20 1/5 = 0.20 1/5 = 0.20
Phenotype frequency: A B AB O	 2/5 = 0.40 1/5 = 0.20 1/5 = 0.20 1/5 = 0.20
Allele frequency: A B O	 3/10 = 0.30 2/10 = 0.20 5/10 = 0.50

GLOSSARY

A

abscission when parts of plants naturally detach, e.g. dead leaves, ripe fruit

abundance the number of individuals in a population

accommodation the process by which the lens of the eye changes shape to focus light on the retina from objects at different distances

accurate having the true value

action potential a change in the electrical potential (membrane potential) of an axon as an impulse (electrical signal) travels along the axon

active acquired immunity immunity developed naturally or artificially through a person being exposed to a live pathogen and developing a primary immune response (developing antibodies)

adaptation a characteristic (or genetic change) that an organism possesses that increases the survival and reproductive chances of that organism in its environment

adaptive immunity also known as the third line of defence, involves the reactions of lymphocytes to the presence of a pathogen

adipose tissue fat tissue

adrenal cortex the outer portion of the adrenal gland

adrenal gland gland situated above the kidney and composed of two parts – the medulla and the cortex

adrenal medulla the inner region of the adrenal gland

adult stem cell pre-specialised cell that occurs in almost every type of tissue, and has lost the ability to divide by mitosis

aerobe only able to live and reproduce in an atmosphere that contains free oxygen

agent a pathogen that causes disease

age-standardised rate an incidence measure calculated as if the population had a standard age structure

agriculture the science and practice of farming plants and livestock

aldosterone hormone produced by the adrenal cortex that increases the reabsorption of sodium ions and decreases the reabsorption of potassium ions in the kidney

alimentary canal the hollow tube along which food passes from the mouth to the anus, including the oesophagus, stomach and intestines

allele variant of a gene

allele frequency a measure of how common an allele is in a population

alternation of generations the alternation of sexual and asexual reproduction as a normal part of a plant's life cycle; involves a sexually reproducing, gamete-bearing generation alternating with an asexually reproducing spore-bearing generation

ambient relating to the surrounding environment (e.g. ambient temperature)

amoeba a single-celled organism that can change its shape by extending and retracting parts of its cell membrane

amylase an enzyme in saliva that converts glycogen and starch to simple sugars

anaemia a condition caused by deficiency of iron in the diet, resulting in pale skin, weakness, unusual tiredness, apathy, low resistance to cold temperatures and difficulty breathing when exerting the body

anaerobe (facultative anaerobe) able to live and reproduce in an atmosphere containing no oxygen

analytical study the statistical analysis of data to test a specific hypothesis

androgens male hormones that control the development and functioning of the male sex organs and secondary sex characteristics

aneuploidy when one or more extra copies of an entire chromosome are made or an entire chromosome is missing, leading to an abnormal number of chromosomes in the cell

angiogenesis the development of new blood vessels

animal husbandry the science of breeding and caring for farm animals

anorexia nervosa severe undernutrition characterised by psychological disorders, excessive weight loss and a distorted body image

anterior the front area of a structure

anthelmintic a class of antiparasitic drugs used to treat internal parasites such as flukes, roundworms and tapeworms

anther the top part of a stamen, the male reproductive organ in a flower; produces pollen

anthropological genetics branch of science that combines components of population genetics with historical, archaeological and linguistic evidence to determine the pathways of human evolution

antibiotics a group of substances that kill bacteria or slow their growth

antibody (immunoglobulin) a type of blood protein produced by the immune system in response to a specific pathogen; its function is to neutralise the pathogen

anticodon three unpaired bases at one end of tRNA that attach the tRNA to its complementary bases on the mRNA strand

antigen a molecule capable of inducing an immune response

antimicrobial agent a substance that kills microorganisms or stops them from reproducing, e.g. antibiotics, antivirals, antifungals, antiprotozoals

antiviral medication medication used to control viral infections; it does not kill the virus but inhibits its development

anvil (incus) the middle bone of the ossicles in the middle ear that is fused with, and transmits sound vibrations to, the stirrup

apomixis a form of asexual reproduction in plants in which new plantlets are produced, often on leaves, without fertilisation or the involvement of a seed

apoptosis programmed cell death

aqueous humour a clear, runny fluid between the cornea and the lens of the eye

array comparative genomic hybridisation (aCGH) a genetic engineering technique used to test a single cell taken from an embryo

artificial insemination a technique whereby sperm are transferred from a male to the vagina of a female without copulation to introduce desirable characteristics from one male to the offspring of one or more females

artificial pollination the deliberate transfer of pollen from the anther of a flower on one plant to the stigma of a flower on the same or another plant, to ensure desirable characteristics are selected

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asbestosis disease caused by the inhalation of asbestos fibres causing an inflammatory reaction in the lung tissue, leading to scarring and stiffening of lung tissue. This makes the process of breathing much harder, and decreases oxygen uptake into the blood

ascertainment bias a form of information bias in which all members of the study group are not followed up to the same extent

asexually not involving the fusion of gametes

asexual reproduction reproduction involving only one parent and no sex cells or gametes, resulting in offspring that are genetically identical to each other and to the parent

assimilation the conversion of simple molecules into more complex molecules that then become part of the structure of cells

ataxia lack of muscle coordination, leading to difficulty in walking

atherosclerosis 'hardening of the arteries', in which fatty deposits on the inner walls of the arteries hinder blood flow

auditory nerve nerve that carries electrochemical impulses from receptor cells in the cochlea to the brain

autoclave a chamber that uses heat and high pressure to sterilise equipment exposed to potential pathogens

autoimmune disease a disease in which the body produces antibodies against its own tissues

autonomic the involuntary branch of the nervous system

autosomal dominant inheritance a pattern of inheritance in which an affected individual has one copy of a mutant gene and one normal gene on a pair of autosomal chromosomes and the mutant gene is expressed

autosomal recessive inheritance a pattern of inheritance in which an affected individual must have two copies of a mutant gene for it to be expressed

axillary bud an embryonic shoot located in the axil of a leaf, which has the potential to form branches

axon single, very long extension of the cytoplasm of the cell body that conduct messages away from the cell body; axons form the 'white matter' of the CNS

B

bacteraemia the presence of bacteria in the blood

bacteriophage a virus that infects bacteria and reproduces inside them

bar graph also known as a column graph; a type of graph used when items have been grouped into unrelated categories; the columns do not touch

barrier an obstacle that prevents the entry of a substance or object

basal plate a modified stem reduced to a short disc

base substitution when one nucleotide base is replaced by a different base, which may result in a different amino acid being inserted into the polypeptide

basilar membrane a membrane below the organ of Corti that pushes up onto the organ of Corti when a sound wave moves through the perilymph

behavioural relating to the way an organism acts

benefit anything that provides an advantage

benign tumour a non-cancerous tumour whereby the cancer cells remain within the boundary of the tumour and do not spread to other body tissues

beriberi disease resulting from prolonged deficiency of vitamin B1 in the diet of children; results in retarded growth, weakened heart muscle, loss of appetite, confusion, inflammation of the nerves, poor coordination, tingling and paralysis

bi-allelic describes a gene that has two variants or two possible alleles within a population (e.g. T and t for height)

binary fission an asexual reproduction process that involves the division of a eukaryotic cell into two, typical of unicellular organisms such as bacteria

bioactive compound a compound that has a biological effect

biodiversity the number, relative abundance and genetic diversity or organisms in an area or on Earth

bioethics refers to the study and investigation of how decisions in medicine and science affect society and the environment

biofilm a slimy layer formed by a group of microbes adhering to each other and to a surface

bioinformatics the science of collecting and analysing complex biological data such as genetic codes using software tools for understanding biological data; a mix of science, statistics, computer science, biology, mathematics and engineering

biolistics the technique of mechanically delivering DNA on microscopic particles into target tissues and cells by 'firing' them from a gene 'gun'

biological fitness the measure of an individual's reproductive success, calculated as the average contribution to the gene pool made by a certain genotype within a population and the relative likelihood that those variants of a gene will be represented in future generations

biological mutagen a mutation agent in the form of a virus or bacterium or some other living organism

biotechnology the use of biological materials as tools

bivalent a homologous pair of chromosomes aligned in early meiosis that form synapses or chiasmata for crossing over

bladder the storage organ for urine before it leaves the body

bleached describes the state of opsin and retinal molecules when they dissociate so that no further light stimuli can be detected

blind spot an area on the back of the eye where all the nerve fibres leave the eye and converge to form the optic nerve

blood-brain barrier a semi-permeable membrane that separates circulating blood from the fluid that bathes the central nervous system

bone conduction implant an implanted microphone that converts sound into vibrations in the skull that are transferred directly to the inner ear by bone conduction, bypassing the outer and middle ear; this technology is used to assist in conductive hearing disorders

bottleneck effect genetic drift that occurs as a result of a natural disaster

Bowman's capsule a spherical, hollow, cup-like structure in the nephron of the kidney that receives filtrate from the glomerulus

bradykinin a compound (peptide) released into the blood to cause smooth muscle to contract and blood vessels to dilate

Bt cotton a transgenic insect-resistant plant genetically engineered by Monsanto; the plant produces a protein that is toxic to bollworm

budding an asexual reproductive process where part of adult organism divides by mitosis and produces a small bud, which separates from the parent and grows into a new individual

bulimia nervosa a condition caused by psychological, environmental and cultural factors where the sufferer is fixated on body image and demonstrates abnormal eating behaviours, including binge eating followed by self-induced purging

burden the number of individual parasites carried by a host

bush medicine traditional medicine; the knowledge and skills used to apply remedies, usually made from plants

C

calyces cup-shaped cavities in the kidney through which urine from the collecting ducts flows before collecting in the pelvis

campaign a range of strategies used to try to reduce the incidence of disease

capsid a protein shell that protects a virus

carcinogenic cancer-causing

carcinoma a cancer that forms in the epithelial tissue, such as the skin or the tissue that lines or covers the internal organs

cardiac arrest heart failure resulting in loss of heart function, breathing and consciousness

carrier an individual who is not affected by a defective allele, but may pass it on to his or her children; an organism that is infected with a pathogen but shows no external signs

case-control study study based on a comparison of individuals or subjects where one group has the disease and the other group (control group) does not

cataract clouding of the lens in the eye, reducing the transmission of light

cell body part of the nerve cell that contains the nucleus, cytoplasm and many organelles

cell cycle a repetitive sequence of cell division and enlargement, including mitosis

cell junction and intercellular 'bridge' formed by the plasma membranes of adjacent cells, enabling contact between the cells

cell-mediated response an immune response involving cytotoxic T cells that eliminates intracellular pathogens

cellulose a substance that makes up plant cell walls; a complex carbohydrate

central nervous system the brain and spinal cord

central nervous system cancer cancer that originates in the brain or spinal cord

centromere the part of a chromosome that holds together newly formed sister chromatids

cerebral haemorrhage leakage of blood into the brain tissue

cerebrovascular disease atherosclerosis of the cerebral arteries

checkpoint inhibitor a type of drug that blocks normal proteins on cancer cells and acts as a type of 'off switch' that helps keep the T cells from attacking other cells in the body

chemoreceptor a cell or organ that detects the concentration of certain chemicals inside the body

chemotactic factor a chemical that causes chemotaxis

chemotaxis the movement of substances or cells in response to a chemical signal

chemotherapy the use of chemical substances or pharmaceuticals to treat disease

chiasmata the points at which the arms of a bivalent meet when chromatids wrap around each other during crossing over

chitin a fibrous substance that is the major constituent of fungal cell walls

choroid the middle layer of the eye; the rear portion is black to reduce the scattering or reflection of light in the eye

chromatid one copy of a newly copied chromosome that is still joined to the other copy by a single centromere

chromatin a complex of macromolecules found in non-dividing cells, consisting of DNA, protein and RNA

chromosomal deletion a mutation that arises when a section of DNA is removed and not replaced, leading to a decrease in the number of genes on a chromosome

chromosomal insertion (duplication) a mutation that arises when a portion of DNA is duplicated and inserted into a chromosome, increasing the number of genes on the chromosome – its effect depends on the size, location and number of repeats

chromosomal inversion a mutation where the reverse order of a sequence of bases arises in a chromosome, caused by a section of DNA being removed, turned around and then re-inserted into the chromosome (usually during replication)

chromosomal mutation (chromosomal aberration) a large-scale change in genetic material where either the overall structure of the chromosome is changed, or the entire number of chromosomes in a cell is altered

chromosomal translocation a mutation that arises when a section of DNA is moved from one chromosome to a non-homologous chromosome (and may involve gene fusion)

chromosome a DNA molecule containing the genetic material that codes the inherited characteristics of an organism

ciliary body a circular structure in the eye, consisting of ciliary muscles and suspensory ligaments

ciliate any single-celled organism with hair-like structures on its surface for locomotion and food gathering

cladode photosynthetic stem

cleavage the division of cells in the early embryo, or in unicellular organisms and protists undergoing binary fission

cloaca a single common opening for the products from the digestive, reproductive and urinary tracts of all vertebrate classes except mammals

cloning making an exact or identical copy

clostridial bacteria a class of bacteria that are Gram-positive and spore forming, e.g. tetanus

clot a thick mass of coagulated liquid, often blood; also called a thrombus

cochlea fluid-filled inner part of the ear; contains receptors that convert sound waves into electrical impulses

cochlear implant hearing technology that converts sound waves into electrical impulses sent to an electrode array implanted in the cochlea, stimulating the nerve endings

coding DNA DNA that codes for products such as proteins

coding strand (antisense strand) the strand in a DNA molecule that is not used as a template during transcription, but its base sequence corresponds to the base sequence of the mRNA transcript produced (but with thymine replaced by uracil); contains codons

codominance a condition where both alleles in a hybrid are expressed (neither allele is dominant or recessive)

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codon a triplet or sequence of three bases on DNA or mRNA that codes for a single amino acid

coenzyme an organic compound, such as a vitamin, which binds with an enzyme

cohort study a study of a group of individuals (cohort) over a period of time to see who develops a particular disease

collecting duct the last site of the tubules of the nephron; contains water, and nitrogenous and other wastes that remain after the processes of filtration, reabsorption and secretion

colonisation resistance the means by which the normal intestinal bacteria protect themselves from colonisation by pathogens

column graph *see* bar graph

commensal relating to or showing commensalism, whereby the organism benefits from another organism without affecting it

communicable disease a disease that can be transferred from one organism to another

community hygiene measures taken to prevent the build-up of pathogens in a community, e.g. sewage and garbage disposal

complement system a group of plasma proteins in the blood that are activated by antibodies or pathogens and enhance (complement) the immune response

concave lens lens that is thinner at the centre and thicker at the edges; used to correct myopia

concurrent happening at the same time

conductive hearing loss hearing loss due to a problem with the mechanical conduction of vibrations from the outer and middle ear to the cochlea

cones photoreceptors in the retina that detect colour

confounding factor, confounding variable variable that influences both the dependent and independent variables

congenital present at birth

conjunctiva the mucous membrane that covers the front of the eye and lines the inside of the eyelids

contact lenses corrective lenses used instead of spectacles and worn directly in contact with the eye

continuity of species the ongoing survival of species as a result of characteristics being passed from parents to offspring in a continuous lineage

continuous breeder an organism that is sexually active all year and in which female fertility occurs in a repeating cycle throughout the year; humans are an example

continuous variation variation within a population where a trait is distributed on a continuum and extends from one extreme to another (rather than increasing in discrete units); an example is height

control centre a region of the brain (e.g. the hypothalamus) that maintains homeostasis by receiving messages from receptors and sending messages to effectors to respond to and counteract a stimulus until the set point is re-established

controlled kept constant

controlled variable a variable that is kept constant so it does not interfere with the outcome of the experiment

convex lens lens that is thicker at the centre than at the edges; used to correct hyperopia

cornea the transparent structure of the sclera that covers the iris, pupil and anterior chamber of the eye

coronary related to the heart

corpus albicans a mass of fibrous tissue in the ovary formed by the degeneration of the corpus luteum if the egg is not fertilised

corpus luteum the Latin term for 'yellow body', which refers to the large mass of vacuolated cells made up of lutein, in a follicle of the ovary

cortex (renal) outer region of the kidney

corticosteroids (glucocorticoids) a class of steroid hormones that are sometimes used to treat inflammation

corticosterone hormone that works with cortisol to regulate the immune response and suppress inflammatory reactions

countercurrent exchange exchange of heat from the warm blood in the arteries (flowing from the heart towards the extremities) to the cooler blood in the veins (coming back from the cold extremities), warming this blood before it returns to the heart

Cri du chat a rare genetic disorder that results from the deletion of a specific section of chromosome 5

cross-infection the transfer of pathogens between people, animals and equipment

CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) a genome editing technique where genes can be spliced and inserted with precision accuracy

crossing over (synapsis) the process during which the arms of a bivalent wrap around each other

culture patterns of human behaviour, such as language, customs, beliefs and values

cystic fibrosis an inherited disease caused by a mutation of the cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7

cystitis inflammation of the bladder's internal lining

cytokines a group of proteins secreted by certain cells of the immune system; play a role in cell signalling by affecting nearby cells

cytokinesis the process following mitosis, during which the cytoplasm divides, separating the daughter nuclei from one another

D

damage-associated molecular patterns (DAMPs) molecules that are released when tissue is damaged and stimulate the inflammatory response

daughter chromosome a chromatid that has separated from its sister chromatid during cell division

deforestation the clearing of a wide area of trees

deletion loss of genes from a chromosome

dendrite an extension of the cytoplasm of a nerve cell body; receives messages in the form of impulses from other axons and conducts these nerve impulses towards the cell body; form the 'grey matter' of the CNS

dendritic cell an antigen-presenting cell of the immune system

dendron a single, elongated dendrite found in sensory neurons

dependent variable a variable that changes during an experiment as a result of the experiment; it is the observed or measured outcome that depends on other factors that have been changed in the experiment

depth study an investigation or an activity completed by a student or students to explore more deeply a topic from the Year 12 Biology course that they find interesting

derived data data that is deduced from raw data by mathematical manipulation, such as graphs, algebraic equations and geometric constructions

dermatophyte fungal pathogen that lives on the skin, nail or hair

dermis the deeper layer of skin between the epidermis and the subcutaneous tissue or hypodermis

descriptive study a study based on describing characteristics, trends and patterns without making a causal link

desiccation a state of extreme dryness

dialysate the fluid that passes through an artificial kidney that has the same concentration as blood plasma without the metabolic wastes

diarrhoea loose watery stools that are passed more frequently than usual

diploid having two sets of chromosomes, or double the haploid number of chromosomes, within any cell

discontinuous variation variation within a population that falls into discrete categories; an example is 'pigment or no pigment'

disease any condition that impairs the normal functioning of a living thing

distal tubule tubule in the kidney, after the loop of Henle, where all required substances are reabsorbed into the bloodstream

distribution where the population of a species is spread within an ecosystem

DNA deoxyribonucleic acid; the hereditary material of an organism in the form of a molecule that carries the genetic instructions used in growth, development, functioning and reproduction

DNA mismatch repair the process of a repair enzyme recognising a mismatched base pair during DNA replication, then excising the incorrect base and replacing it with the correct base

DNA polymerase III an enzyme that adds DNA nucleotides during DNA replication

DNA profiling (DNA fingerprint analysis) a technique used to identify and compare individuals based on characteristics in their DNA

DNA primase an enzyme that catalyses the synthesis of RNA segments called primers

DNA repair gene a gene involved in DNA repair, for example by coding for proteins that stop the cell cycle while other proteins remove damaged regions of DNA and replace them with the correct sequence

DNA sequencing the process of determining the exact nucleotide sequence (order of bases A, T, G and C) of a gene on a chromosome

domain one of three main categories (eukaryotes, bacteria, archaea) into which all living organisms are currently classified

dominant describes a trait that is expressed (appears) in a heterozygous individual

dysphagia difficulty with swallowing

dysphonia inability to speak (or bark, in dogs)

E

ectoparasite a parasitic organism that lives on the outside of the host organism

educational program program that educates the public about the effects of a disease, risk factors and strategies to prevent the disease

effector muscle or gland that carries out a response

efficacy the degree to which a treatments produces the desired result

electrochemical impulse the form in which messages are transmitted through nerves; made up of electrical impulses and chemical messengers

electromagnetic radiation a form of energy that comes from the sun and is found all around us, such as radio waves, microwaves and gamma rays

electromagnetic spectrum (EM spectrum) the overall range of wavelengths of electromagnetic radiation

electronic cigarette (e-cigarette) a battery-powered device that forms an aerosol that is inhaled into a person's lungs

electroporation the use of an electrical current to increase the permeability of the nuclear membrane temporarily, to allow DNA to enter the nucleus

e-liquid liquid in an e-cigarette that is changed into an aerosol

embryonic diapause a delay in the development of an embryo until conditions are favourable, as a strategy to increase the chances of survival of the young

embryonic stem cell cell separated from the embryo and cultured separately

emesis vomiting

empirical able to be verified by evidence gained through observation and experimentation

endocrine gland gland that secretes hormones directly into the bloodstream

endocytosis the taking in of molecules by a cell, by making a fold in the cell membrane and forming a vacuole

endogenous having an internal cause or origin

endogenous pyrogen a molecule produced by leukocytes in response to pathogens that increases the set point for body temperature, inducing a fever

endometrium the lining of the uterus

endoparasite a parasitic organism that lives within the host organism

endospore a tough structure surrounding some bacteria that protects the bacterium from environmental stresses during dormant periods

endothelium, endothelial cells the single layer of cells that line organs and cavities in the body, such as blood vessels, heart, lymphatic vessels

endotherm an organism that maintains body temperature within a narrow range of tolerance limits despite variations in the ambient temperature

endotoxin a toxin released by a bacterial cell when it disintegrates

enucleation removal of the nucleus of a cell

enzootic endemic in an animal population; regularly infecting animals in a certain region

enzyme a biological catalyst that increases the rate of a chemical reaction in the body

epidemic the widespread occurrence of an infectious disease in a community at a particular time

epidemiologist a person who studies disease in a population (as opposed to someone who studies disease in an individual organism)

epidemiology the study of the distribution and determinants of health-related states or events (including disease) and the application of this study to the control of diseases and other health problems

epidermis the outer layer of skin cells

epigenetics changes in an organism caused by changes to gene expression rather than alteration of the genetic code itself

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epithelial tissue, epithelium a thin tissue that forms the outer surface of the body and the inner lining of the digestive system and other hollow structures

epitope the part of an antigen that is recognised by the immune system

epizootic a disease of the animal (non-human) population

euploidy where there is a gain of one or more complete sets of chromosomes, resulting in a chromosome number that is an exact multiple of the haploid number

eutherian a placental mammal; the young complete their embryonic development inside the uterus of the mother, following internal fertilisation

evaporative cooling a means of cooling the body, whereby water evaporating from the body removes heat

exon a sequence of bases in DNA and in mature mRNA that is translated into proteins, once introns have been removed from the initial pre-mRNA transcript

exotic not native to a certain region

experimental study a study in which individuals are randomly allocated to groups and the investigator decides which group is to receive the intervention

external fertilisation fertilisation (uniting of the nuclei of a sperm and an egg) that occurs outside the body

extracellular space the fluid that bathes the outside of cells

F

faecal egg count also called a worm test; used to estimate the parasite burden in an animal by counting eggs per gram of dung

falsifiable able to be disproved

feedlot a form of primary industry where cattle are fattened before slaughter

fermentation the process that results from the use of the biological organism yeast

fertilisation the process that occurs when the haploid nucleus of the egg fuses with that of the sperm, forming a fertilised egg called a zygote

fibrin a fibrous protein involved in clotting

fibrous proteins proteins that form a long chain, are insoluble in water and a structural component of protoplasm in cells and of tissues

fidelity of replication the exact copying of the sequence of bases in DNA during replication to ensure the accuracy of heredity and gene expression

filtration a process in the kidney whereby only substances that are small enough pass through the walls of the capillaries of the glomerulus into the Bowman's capsule, while larger molecules remain in the capillaries

firewall in computing, a system designed to limit the spread of a computer virus

flagellate any single-celled organism with a whip-like tail, e.g. *Euglena*

follicle stimulating hormone (FSH) a reproductive hormone that causes sperm production in men and follicle development in women, causing an egg to mature in an ovary; it also stimulates ovaries to secrete oestrogen

fomite an object or material that is able to carry infective pathogens, e.g. surfaces, clothes, bedding

founder effect genetic drift that occurs due to a few individuals in a population becoming geographically isolated from the original population

fovea a depression in the macula of the eye that contains densely packed cones and is the area of greatest visual acuity

frameshift mutation a mutation in which the reading frame of mRNA is shifted, so that triplets of bases that normally form codons are no longer grouped together, resulting in a gene being translated unnaturally from the position of the mutation onwards

fruit the fleshy part of a plant that contains seeds and develops from the ovary

G

gamete special sex cell that carries genetic information from both parents to the offspring

gametogenesis the process in which gametes are produced in the reproductive organs

gastroenteritis inflammation of the stomach and intestines; causes diarrhoea and vomiting

gene the basic unit of heredity; each gene consists of a segment of DNA with nucleotide bases in a specific sequence, and forms part of a chromosome

gene cloning the process of producing identical copies of a gene

gene expression the conversion of DNA into a product such as protein; the genetic instructions given to a cell to create its structure and ensure its correct functioning

gene flow when new individuals enter a population, or existing individuals leave, which may result in a change in allele frequency

gene mutation a mutation that affects only a single gene

gene pool all the alleles of all the genes in a breeding population

gene regulation the mechanism controlling the 'switching on' and 'switching off' of genes to increase and decrease the production of gene products (protein or RNA)

gene technology the manipulation of genetic material (DNA) to create products for human use

gene therapy the transfer of a normal gene into an individual to correct a genetic disorder

genetic disease a disease caused by mutation of a gene or a chromosome

genetic diversity the total of all the genetic characteristics in the genetic make-up of a species

genetic drift a change in allele frequency due to random chance

genetic engineering the intentional manipulation of genetic material to alter the genetic makeup of an organism

genetic marker an identified sequence of DNA at a known site on a chromosome

genetic recombination genetic variation caused by crossing over and random segregation in meiosis and recombination of paternal and maternal genes within each gamete

genetic stability the consistent passing on of genetic information from parent to offspring, with little or no change, over time, in the sequence of genes in chromosomes or of nucleotides in DNA

genetic variability the tendency of individual genetic traits in a population to vary

genetic variation differences in various genetically determined traits or features among members of a population

genetically modified organisms (GMOs) organisms that have DNA from more than one species

genome the complete set of genetic material within a cell or that an organism has in each of its cells

genome-wide association study (GWAS) a study that involves using computer software to rapidly scan identified genetic markers across complete sets of DNA, or genomes, of many people to find genetic variations associated with a particular disease

genomics the field of study concerned with sequencing the nucleotides of complete sets of genes in organisms

genotype the combination of genes that an organism has

genotyping the identification of genetic variations in individuals

genus a classification category between family and species

germline cell a cell that gives rise to gametes

germline mutation (gametic mutation) a mutation that occurs in the sexual reproductive cells (germline cells) that give rise to gametes and can be passed to every cell in the offspring

germplasm any living tissue from which new plants can form, including whole plant parts, pollen and clusters of cells

globular protein a protein, usually spherical, compact and soluble in water; often transports proteins, such as haemoglobin in blood

glomerular filtrate the substance that passes through the walls of the capillaries of the glomerulus in the kidney and moves into the Bowman's capsule

glomerulus a spherical network of capillaries inside the Bowman's capsule of the kidney

glucagon a hormone produced by alpha cells in the pancreas; stimulates the production of glucose

goitre enlargement of the thyroid gland resulting in a visible lump in the throat; caused by prolonged iodine deficiency

gonad a male or female reproductive organ that produces gametes

gonadotropic hormones (gonadoptropins) hormones produced in the hypothalamus and pituitary gland that stimulate the gonads, or sex glands, to carry out their reproductive or endocrine functions

Graafian follicle an enlarged, dominant follicle that pushes to the surface of the ovary and creates a bulge during the follicular phase

Gram-positive/negative classification of bacteria into two categories, based on the way a sample looks under the microscope when stained by the Gram stain technique; the stain differentiates between the cell walls of certain bacteria

granuloma a 'wall' formed by immune cells to separate a pathogen from body tissues

granzymes chemicals that cause lethal damage to bacteria; found in cytoplasmic granules of certain lymphocytes

H

haemodialysis a process in which blood is removed from the body and passed through an artificial kidney to remove urea and other wastes

hair cells receptor cells in the organ of Corti, in the inner ear, that convert the kinetic energy of a sound wave into a nerve impulse when they are bent against the tectorial membrane by pressure from the basilar membrane

hammer (malleus) the first of the three ossicles in the middle ear; touches the eardrum and transfers vibrations to the anvil

haploid having one complete set of chromosomes within a cell

haplotype a group of genes that are inherited together on a particular chromosome from one parent

haplotype network a diagrammatic representation of the genetic diversity within a group

Hardy–Weinberg principle a principle based on the idea that the frequency of alleles in a population remains constant from one generation to the next if none of the evolutionary influences (e.g. mutation, sexual selection, genetic drift, gene flow) are acting

healthy worker bias a form of selection bias in which participants in a study are generally healthier than those who do not participate

hearing aid a form of technology used to magnify sound vibrations to better enable their transmission to the middle and inner ear

helicase an enzyme that causes the DNA helix to unwind and the strands to separate, to allow for replication

helminth a parasitic worm

herd immunity a form of protection given to individuals in a population, both vaccinated and unvaccinated, against infectious disease when a significant proportion of the population has been vaccinated

heredity the passing of characteristics or traits genetically from one generation to the next

hermaphrodite in animals, having both male and female reproductive organs on the same individual, sometimes termed bisexual

heterozygous hybrid

hibernation an extended period of inactivity in response to cold, where the body temperature does not drop below 30°C, but the heart rate and oxygen consumption drop considerably

hilum the curved indentation of the kidney where the blood vessels enter and leave the kidney and the ureter leaves the kidney

histamine a nitrogenous compound involved in allergic and inflammatory reactions; causes smooth muscle to contract and capillaries to dilate

histogram a graph similar to a column graph but which has columns that do touch each other because they represent continuous numerical data

histone a protein found in eukaryotic cell nuclei that packages and orders the DNA into structural units

histopathology the study of changes in tissues caused by disease or injury

homeostasis the maintenance by an organism of a relatively constant internal state, regardless of external changes in the environment

homologous pair a pair of chromosomes in a cell (two chromosomes and four chromatids), with one chromosome coming from each parent, where the two chromosomes carry alleles for the same genes

homozygous having identical alleles of a particular gene in a diploid cell, for any particular hereditary characteristic (e.g. TT or tt)

horizontal transmission viral spread through individuals of the same generation

hormone a chemical substance that acts as a messenger in the body, coordinating many different aspects of functioning, including metabolism and reproduction, so that actions within the body are synchronised

human papilloma virus (HPV) a virus linked to cervical and other cancers

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humoral response an immune response involving the release of antibodies by plasma cells

hybrid vigour the result of stronger, healthier offspring due to cross-breeding with different varieties of organisms

hydrocortisone (cortisol) a hormone that regulates how the body manages stress and how it converts carbohydrates, fats and proteins to energy; also plays a role in regulating cardiovascular function and blood pressure

hyperopia an eye condition in which distant objects are seen clearly, but near objects are out of focus; longsightedness

hypertension high blood pressure

hyphae (sing. hypha) the branching filaments that make up the mycelium of a multicellular fungus, such as mould

hypodermis the tissue beneath the dermis of the skin

hypothalamus a region in the lower central part of the brain that is an important control centre for maintaining homeostasis

hypothesis a tentative prediction or explanation of an observation, usually based on an existing model or theory

I

immune system the organs and tissues that provide resistance to infection

immunisation the process by which an individual's immune system is prepared to challenge a specific pathogen in advance of exposure, making a person immune to a disease

immunoglobulin any protein present in the blood that functions as an antibody

immunoglobulin A (IgA) an antibody found on the mucus membranes that protects them against infection

immunotherapy treatment that uses certain parts of the immune system to help protect against infection or disease

inbreeding the breeding of closely related organisms over many generations

incidence (of disease) the probability of occurrence of a disease in a population at any one time

inclusion bodies non-living particles inside a cell, commonly virus particles in cytoplasm; used to diagnose some viral infections

incomplete dominance a condition in which a gene is expressed as a blending of the characteristics of the two alleles present in a hybrid (neither allele is dominant or recessive)

incubation period the time that elapses between exposure of a person to a pathogen and when symptoms first become apparent

independent variable the variable that is controlled or manipulated by the experimenter

induced mutation a mutation caused by a mutagen (a chemical or biological agent or some form of radiation that causes a change in DNA structure)

infectious disease any disease caused by a pathogenic organism

inflammation a reaction to the presence of an antigen, in which tissues become hot, red, swollen and painful

influencers individuals or companies on social media who can drive conversations and influence others

information bias errors in recording measurements or information; affects the validity of research results

innate immunity the non-specific immune system, including the first two lines of defence (e.g. physical barriers to entry, phagocytes, inflammation)

inner segment part of a rod or cone (receptor cell) in the eye that connects the outer segment of the cell to the conducting part of the cell

insulin hormone produced by beta cells that causes glucose in the blood to be removed

intellectual property a created work where the work's creator has the sole right to make a commercial profit from it

interferon a signalling protein released in response to the presence of a pathogen, particularly viruses

interleukin a protein involved in the proliferation of lymphocytes during an immune response

internal fertilisation fertilisation (uniting of the nuclei of a sperm and an egg) that occurs inside the body

interneuron a neuron within the CNS that transmits signals between sensory and motor neurons

interoceptor a receptor that detects internal stimuli related to homeostasis

interphase a stage of the cell cycle that precedes mitosis, during which the cell prepares for division

intervention study a study based on comparing individuals or subjects, where one group has received the intervention, and the other group has not

interviewer bias a form of information bias in which the interviewer indirectly leads study participants to reply to questions in a certain way

intraocular lens implantation (IOL) the removal of a cloudy lens and the insertion of an artificial lens

intravascular space the fluid inside blood vessels

intron a sequence of bases that is removed from pre-RNA after transcription, so a continuous sequence of coding bases remains in mature RNA for polypeptide production; also the corresponding non-coding nucleotide sequences in DNA

in vitro fertilisation (IVF) a process in which an egg is fertilised by sperm outside the body

iodopsins the three different types of visual pigments responsible for detecting colour; each cone contains one iodopsin

ion an electrically charged atom

ionising radiation a harmful type of radiation, which has enough energy to break chemical bonds in molecules, including DNA

iris a ring of muscle that forms the coloured part of the eye and controls the size of the pupil to regulate the amount of light entering the eye

ischaemic heart disease atherosclerosis of the coronary arteries

islets of Langerhans groups of cells that make up the endocrine portion of the pancreas

isoproteins proteins that have a similar structure and/or function but are not identical; produced through the alternative splicing of mRNA

K

kidney an organ containing millions of nephrons, responsible for the excretion of urea and osmoregulation; made up of three main regions: cortex, medulla and pelvis

Koch's postulates criteria for establishing a causative relationship between a microorganism and a disease

kwashiorkor a disease caused by a severe lack of protein in the diet and resulting in failure to grow, enlarged liver, hair changes, apathy, irritability and increased susceptibility to infectious diseases

L

lacrimation flow of tears

lactogenic hormone hormone secreted by the pituitary gland in females; acts on breast tissue to prepare for and maintain milk production

lagging strand the strand in a DNA molecule on which nucleotides are added in chunks, called Okazaki fragments, from the replication fork backwards, and in which replication is discontinuous, with fragments being joined by ligase to form a continuous strand

lameness difficulty walking due to injury to a leg or foot

LASIK surgery a surgical technique that uses lasers to change the curvature of the cornea to correct myopia and hyperopia

leading strand the strand in a DNA molecule in which replication is continuous, and on which nucleotides are added in a long chain, growing in the same direction as the replication fork opens up

legislation laws made by parliament

lens a biconvex, transparent, highly elastic structure in the eye that changes shape to refract light

lens capsule a clear, membrane-like, elastic structure that encloses the lens of the eye

lesion any abnormal change or damage to a tissue or an organ

leukaemia a cancer that forms in the bone marrow and other blood-forming tissues

levy a tax imposed by the government

Leydig cells cells in the seminiferous tubules of the testes that make testosterone

lifestyle disease a disease that is a direct result of the way in which individuals live their lives

ligand a protein that attaches (binds) to a receptor protein that has a specific complementary site into which the ligand fits

ligase the enzyme that seals together the two strands of newly replicated DNA

ligation the rejoining of pieces of DNA using the enzyme DNA ligase

limit of reading the smallest unit of measurement on a measuring instrument

line graph a type of graph used to show the relationship between continuous variables

line of best fit a straight line that is fitted to a graph of data points

literature review a report and evaluation of information from secondary sources on a topic of interest

locus (pl. **loci**) a segment of DNA; the position of a gene on a chromosome

logbook the record of an experiment or investigation kept by the scientist performing the experiment; it is a legal record of the experiment and its results

loop of Henle part of the tubule that is in the medulla of the pelvis; plays a role in osmoregulation

loss to follow up bias a form of information bias in which not all subjects who began the study are available at the end of the study

luteinising hormone (LH) a reproductive hormone that causes testosterone production in men and development of the corpus luteum after ovulation, as well as lactation, in women

luteinising phase the phase after ovulation, when the burst follicle in the ovary enlarges and changes colour, building up a yellow protein called lutein

lymphocyte white blood cell involved in the adaptive immune response

lymphoid tissue that is involved in the defence against pathogens, e.g. spleen, thymus, lymph nodes

lymphoma a form of cancer that originates in the lymphatic system and cells of the immune system

lysis destruction of a cell wall or membrane

M

macrophage a large phagocyte found at the site of tissue inflammation

macula an area at the back of the eye, in the centre of the retina, where incoming light is received

major histocompatibility complex (MHC) a set of cell surface proteins required to recognise antigens; found in vertebrates

malignant tumour an abnormal growth of cells (cancer) that is not confined by a boundary and spreads to other tissues in the body

malnutrition an imbalance in the diet that leads to undernutrition or overnutrition

management care and attention given to an individual suffering from a disease

mange skin condition caused by mite infestation

mannan a plant polysaccharide that occurs in some plant cell walls

marasmus a disease in which the sufferer is extremely underweight and has lost most of their subcutaneous fat; they are very weak and susceptible to infection, and if untreated can die from starvation or heart attack

marsupial a mammal whose young develop internally for a short time, then continue their embryonic development in a parental pouch

maternal inherited from the mother (referring to, for example, chromosomes)

mature mRNA the molecule that results from the splicing of mRNA after transcription and the removal of introns

measurand the quantity being measured

measurement bias a form of information bias in which inaccurate measurements are made

medulla the middle region of the kidney

meiosis a type of cell division that halves the chromosome number and gives rise to gametes that transmit genetic material from one generation to the next during sexual reproduction

melanoma a potentially deadly form of skin cancer originating in the melanocytes (melanin-producing cells)

memory T cell T lymphocyte capable of responding to a specific antigen long after primary exposure

menarche the commencement of ovarian and menstrual cycles during puberty

meningitis an inflammation of the membranes (meninges) that cover the brain and spinal cord

menopause the cessation of reproductive capacity in women, triggered by decreased sensitivity of the ovaries to FSH and LH

menses the start of the menstrual cycle, also known as the menstrual period; lasts about four days, during which time the endometrium breaks down and tears away, accompanied by bleeding

menstrual cycle a cycle of changes in the uterus, required for preparation of the uterus for pregnancy

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menstruation bleeding caused by the breaking down and tearing away of the endometrium during menses

meristem tissue in plants that can divide to form any other tissue

mesothelioma a form of cancer that occurs in the membrane surrounding the lungs

messenger RNA (mRNA) an intermediate molecule that carries a transcribed copy of relevant instructions from the nucleus to the ribosomes in the cytoplasm; it is single-stranded and not twisted into a helix

metastasis the spread of cancer cells from the primary site to another part of the body

microbiology a branch of science dealing with microorganisms

microbiome beneficial bacteria that inhabit the human body

micro-injection a process used to deliver DNA into egg cells when creating transgenic species

micturition urination; release of urine from the bladder

misclassification bias a form of information bias in which some subjects are already suffering from the condition but are undiagnosed at the beginning of the study

mispairing the insertion of incorrect nucleotides opposite a template strand of DNA during replication

missense mutation a point mutation that results in an amino acid change; the type of replacing amino acid will determine the functionality of the protein

mitochondrial DNA (mtDNA) a small, circular DNA molecule found in the respiratory organelles (mitochondria) of cells; can be used to trace maternal inheritance

mitosis the part of the cell cycle during which active nuclear division takes place, to ensure that replicated chromosomes separate and are equally distributed to the daughter cells

mitotic index the ratio of the number of cells in a population undergoing mitosis, to the total number of cells in a population

mobility migration within a population

model a representation of a system or phenomenon that explains the system or phenomenon; may be mathematical equations, a computer simulation, a physical object, words or another form

modern synthesis a combination of the concepts of Mendelian genetics and Darwinian evolutionary theory, in the modern understanding of evolution

monoculture the cultivation of a single species of crop in a given area

monogenic describes a disease that is controlled by a single gene

monosomy the condition in which the total number of chromosomes is one less than the normal diploid number

monotreme a mammal that lays eggs rather than bearing live young; examples are platypus and echidna

morbidity the number of people affected by a disease

mortality the number of people who die from a disease

mortality rate the number of deaths due to a particular disease in a specific time period (usually one year)

motor neuron a nerve cell that transfers messages from the CNS (brain or spinal cord) to effectors such as muscles or glands

mucous membrane a membrane that lines an internal cavity in the body

mucus plug a plug that fills and seals the cervix during pregnancy

multi-allelic describe a single-gene trait for which there are three or more alleles, such as in ABO blood groups

multifactorial involving a number of causes or factors

Multiregional hypothesis (MRE) the theory, based mostly on fossil evidence, that all human populations can be traced back to when *Homo erectus* first left Africa, about 2 million years ago

mutagen an environmental agent that alters DNA, causing mutations

mutagenesis the process of inducing a mutation

mutagenic mutation an error in DNA replication arising as a result of the exposure of cells to environmental factors such as radiation or chemicals

mutation an unusual error in DNA replication, involving incorrect base pairing, which leads to a change in the DNA base sequence

mycelium the main fungal body of multicellular fungi; formed from hyphae

mycosis a disease caused by a fungus

myeloma a malignant tumour of the bone marrow

myopia when a person can see near objects clearly, but distant objects appear blurred; shortsightedness

N

natural reservoir a population in which a pathogen naturally lives

naturally occurring mutagen a mutagenic agent that is present at normal levels within natural environments, that may cause mutations

necrosis destruction of tissues

negative feedback mechanism a body system in which a response is initiated to counteract a stimulus and return internal conditions to the set point, maintaining homeostasis

nephron the functional unit of the kidney that removes urea and balances salt and water levels in the blood

neurotoxin a poison that acts on the nervous system

neutral mutation a change in DNA that does not significantly affect the structure or functioning of the protein product

neutrophil the most common type of white blood cell, involved in phagocytosis of pathogens and other small particles

non-biological mutagen a naturally occurring mutagen in the environment that originates from non-living matter, such as metals

non-coding DNA DNA that contains sequences that are excised from RNA and do not code for products such as proteins

non-coding strand (sense strand) a strand of DNA that serves as a template during mRNA transcription; its sequence of bases is the same as those of the anticodons on tRNA (but with thymine replaced by uracil)

non-disjunction incorrect separation of chromosomes during cell division

non-infectious disease disease that is not caused by a pathogen and is not contagious

non-nuclear DNA DNA that is located not in the nucleus but in organelles in eukaryotic cells, such as the mitochondria and chloroplasts

nonsense mutation a mutation that changes an amino acid to a stop codon, with a very noticeable effect on a protein as it cuts it short; the resulting protein is usually non-functional

notifiable describes a category of infectious diseases that must be reported to authorities by law; examples are measles in humans, foot and mouth disease in cattle

nucleoid a dense region in the cytoplasm of prokaryotic cells

nucleotides simple repeating units comprising a simple sugar, a phosphate and a nitrogenous base, which make up nucleic acids, and which may be linked together to form single chains, as in RNA or double strands, as in DNA

O

obligate when an organism is restricted to a particular environment or way of life; for example, an obligate anaerobe cannot survive in an oxygenated environment

observational study study based on collecting qualitative data through observation

occlusion blockage

oedema swelling caused by fluid retention

oestradiol the main form of oestrogen

oestrogens the main group of female hormones; control the development and functioning of the female reproductive system and secondary sex characteristics

oestrus the period of female fertility in animals, commonly referred to as the animal being 'in oestrus', 'in season' or 'on heat'

off-label use the use of a pharmaceutical for an unapproved application, age, dose or route of administration in an animal or plant

oncogene a type of gene that can cause uncontrolled production of cells (cancer) and prevents cell death

opportunistic affecting those who are immune suppressed

opsin a protein that is combined with retinal to form a visual pigment

opsonisation an immune process whereby an antigen is marked for destruction by immune cells (phagocytes)

optic nerve the nerve that carries messages from the receptors in the retina of the eye to the brain

optometrist person who specialises in eye health

organ of Corti a structure in the middle canal of the cochlea containing receptors in the form of hairs that convert vibrations into electrical impulses

osmoreceptor a specialised cell in the hypothalamus that detects changes in osmotic pressure

osmoregulation the maintenance of optimal levels of salt and water in the bloodstream

ossicles the set of small bones in the middle ear that magnify vibrations from the eardrum and transfer these to the oval window

osteomalacia a disease in adults caused by a lack of vitamin D, resulting in skeletal and muscular problems

osteoporosis thinning of the bones

outer segment part of a rod or cone (receptor cell) in the eye that contains the visual pigments

outlier a data point that is distant from the other data points in the sample

oval window a membrane that separates the middle ear from the fluid-filled inner ear (cochlea)

ovarian cycle a series of changes in the ovary of a female mammal, during which the follicle matures, the ovum is shed and the corpus luteum develops

ovary the female reproductive organ where eggs are produced; in flowers, protects the fertilised and developing ovules and grows to become a fruit

oviparous describes animals in which the embryo develops inside an egg but outside the female's body

ovo-viviparous describes animals in which the embryo develops within an egg inside the mother's body, and hatches before being born alive

ovulation the release of an ovum (egg) by a mature follicle in an ovary

ovule a plant structure inside the ovary of a flower, containing an egg cell and developing into a seed when fertilised

oxidative burst the rapid release of reactive forms of oxygen (e.g. hydrogen peroxide) from certain cells

oxytocin a hormone that promotes the coordinated contraction of the smooth muscle of the uterus and the softening of the cervix, so a baby can be born

P

pancreas an organ that includes an endocrine portion that releases the hormones insulin and glucagon

parasitic describes an organism that obtains its needs from another organism while offering no benefit in return

parathyroid gland a small gland embedded in the thyroid gland that releases parathyroid hormone

parathyroid hormone hormone secreted by the parathyroid glands and responsible for increasing the amount of calcium in the blood when levels are too low

parthenogenesis a form of asexual reproduction involving a female parent only, resulting in young that are genetic clones of their mother

passive acquired immunity immunity that develops when antibodies are introduced into the body, without the person being exposed to a pathogen directly

pasteurisation the process of heating a liquid to destroy any disease-causing bacteria

patent a licence issued to the inventor of a novel method or technology; prohibits others (for a specific time) from making or selling the invention

paternal inherited from the father (referring to, for example, chromosomes)

pathogen any organism that causes disease in another

pathogen-associated molecular patterns (PAMPs) molecules associated with certain pathogens that are recognised by the cells of the immune system

pathogenicity the ability of an organism to cause disease

patient zero the initial patient in an infectious disease outbreak

pedigree chart a universally accepted graphical representation, in scientific format, of the inheritance of a particular genetic trait over a number of generations

peptide bond a chemical bond that holds together the amino acids in a linear sequence or polypeptide chain

peptidoglycan the substance that forms the cell walls of many bacteria

perennating organ an underground organ such as a root or stem that contains stored food and buds which will grow if the organ is split, in a form of asexual reproduction, ensuring survival in adverse conditions

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perforin a protein released by killer cells in the immune system that perforate (punch holes in) pathogens

perilymph the fluid in the upper and lower canals of the cochlea

perinatal the time shortly before and after birth

peripheral nervous system all the nerves throughout the body that are not part of the CNS

peritoneal dialysis a process in which wastes in a person's blood are filtered out through the peritoneum of the person's abdomen

personal hygiene steps taken by a person to maintain cleanliness of their body

pesticide a substance used to kill pests; may be an insecticide (kills insects), fungicide (kills fungi) or herbicide (kills weeds)

Peyer's patches lymphoid tissue found in the gut

phagocyte a type of white blood cell that engulfs and digests other small cells and particles

phagocytosis the process by which a phagocyte engulfs and destroys a foreign particle

phenotype the physical appearance of an organism, as determined by its structure, behaviour and physiology

phenylalanine an amino acid found in some proteins

phenylalanine hydroxylase an enzyme required for the first step in the breakdown of the amino acid phenylalanine

phenylketonuria (PKU) a genetic disease caused by mutation of the gene on chromosome 12, which codes for the enzyme phenylalanine hydroxylase (PAH)

pheromone chemical substance released by an organism such as an insect, that has an effect on another organism, including sexual attraction

photophobia sensitivity to light

photoreceptor a receptor cell in the eye that is stimulated by light energy

phyllode photosynthetic leaf stalk

physiological relating to the way an organism's body functions

pie graph a circular graph that shows fractions of a whole; also known as a sector graph

pinna the fleshy part of the outer ear; directs sound waves into the auditory canal

pituitary gland an endocrine gland situated just below the hypothalamus that secretes a number of hormones that regulate other endocrine glands, including ovaries and testes

placebo a treatment with no active property, usually a sugar pill

plaque a hard, calcified substance deposited on the walls of blood vessels that reduces the elasticity of the arteries and blood flow

plasmid a small ring of non-chromosomal DNA that floats separately in the cytoplasm of prokaryotic cells and replicates independently of the chromosome

platelet also called a thrombocyte; a cell that helps in the formation of a clot to stop bleeding

plumule a young stem that grows from a plant embryo and produces green leaves for food production

pluripotent in relation to stem cells, capable of dividing and giving rise to any type of tissue

point mutation a change to a single base pair of DNA, affecting only a single gene

polarised describes the state of a membrane when the inside of the membrane is negative relative to the outside of the membrane

pollen a fine powdery substance comprising pollen grains, produced by the anthers; contain the male gametes of seed plants

pollination the transfer of male gametes (pollen) from the anthers of one plant to the stigma of the same or another flower so that fertilisation may occur

polygenic describes a trait that is coded for by two or more genes, with each gene having its own set of alleles

polymerase chain reaction (PCR) a technique used to make multiple copies of a segment of DNA, in order to increase, or amplify, the amount of DNA available in a sample to be studied

polymorphism when individuals have different phenotypes, usually arising as a result of mutation; when are different forms of the same gene

polypeptide chain a long chain of nitrogen-containing amino acids, of which proteins are made up

polysaccharide capsule an extra slime layer on some bacterial cells that protects them from the environment

population genetics the study of how the gene pool of a population changes over time, leading to a species evolving

posterior back area of a structure

poultice a soft mass of material applied to the body to relieve soreness and swelling

precise what measurements are called when they are close to each other

pre-implantation testing procedure used to test a single cell taken from a three-day-old embryo for mutated genes prior to implantation

premonition protection from severe infection by a pathogen due to a chronic low-grade infection by the same pathogen

pre-mRNA mRNA transcribed from DNA, in eukaryotic cells, before further editing takes place in the nucleus to produce mature mRNA

prenatal during pregnancy; before birth

prevalence (of disease) the proportion of a particular population affected by a disease at any one time

prevalence/incidence bias a form of selection bias in which only current cases are included in a study, excluding those who have died or recovered

prevention efforts to stop the occurrence of disease

primary data data that you have measured or collected yourself

primary immune response the first response of the immune system when exposed to a pathogen

primary industry any industry that gathers raw materials for energy or consumer products, e.g. mining, agriculture

primary lymphoid tissue sites in the lymphatic system where lymphocytes develop, such as bone marrow

primary structure the basic structure of a protein, arranged as polymers of amino acids in linear chains

primary tumour area in the body where the initial malignant tumour develops

primer a short strand of RNA that attaches to the DNA to allow synthesis to be initiated during DNA replication

probiotics microbes that are believed to provide health benefits when ingested

progesterone the most common progestogen; the hormone that plays a primary role in pregnancy and stimulates the secretion of milk in mammary glands

progestin synthetic form of progesterone

progestogen a generic term for a group of female hormones that have a progestogenic effect, e.g. progesterone

prokaryotic describes a unicellular organism that lacks a membrane-bound nucleus

prolactin a lactogenic hormone that acts on breast tissue to prepare for and maintain milk production

promoter a region of DNA that initiates transcription of a particular gene

prophylactically given to prevent infection by a pathogen

prostaglandin a hormone secreted by the wall of the uterus to initiate labour; dilates blood vessels and inhibits platelet aggregation

prosthetic group (cofactor) a non-protein part of an enzyme to which conjugated proteins are attached

protease an enzyme that breaks down proteins and peptides

protein a large molecule made up of polypeptide chains, formed by sequences of amino acids linked together by peptide bonds, and which contains the chemical elements nitrogen, carbon, hydrogen and oxygen

proteomics the large-scale study of sets of proteins produced in an organism or biological system to establish how the proteins in a variety of cell types work together

proto-oncogene a gene that codes for proteins that stimulate cell growth and mitosis

proximal tubule tubule close to the Bowman's capsule in the kidney, through which substances required by the body are reabsorbed into the bloodstream

pseudoscience a belief that is mistakenly regarded as being based on scientific method

pupil the hole in the centre of the iris, where light enters the eye

purebreeding homozygous, such as in a pair of individuals with identical alleles for a gene who will therefore always produce offspring of the same phenotype for that gene

pyrexia-fever when a person's body temperature increases above its set point

pyrogen a substance that produces a fever when released into the blood

Q

quarantine a public health measure based on restricting the movement of people, plants and/or animals through ports of entry to a country, to prevent or limit the spread of pests and contagious diseases

quasi-experimental study an empirical study of cause and effect in a population, without random assignment of subjects

quaternary protein structure the three-dimensional structure of a protein made up of two or more polypeptide chains (folded or pleated) that are linked to create a complex but very specific shape

R

radiation any transfer of energy through space from a source, such as electromagnetic radiation from the sun; (in treatment) the use of radiation to treat a disease

radicle a young root that grows from a plant embryo to absorb water and soil nutrients

random error an error that is unpredictable and has an inconsistent effect on measurement within a study

rate of immunisation the proportion of people within a population who have been vaccinated against a particular disease

raw data original data taken directly from a measurement system

reabsorption the process by which substances required by the body move from the tubules of the kidney back into the capillaries by diffusion or active transport

recall bias a form of information bias in which subjects' ability to recall information varies

receptor a group of sensory cells within the body that detect a change in a particular component of the internal environment

recessive describes a trait that is hidden or masked in a heterozygous individual

recombinant DNA DNA that is made up of DNA from more than one genome

relaxin a hormone that aids the softening of the cervix during childbirth

reliable giving the same results within experimental uncertainty

renal artery an artery carrying blood with a high urea concentration into the kidney

renal capsule a tough, fibrous, fatty layer surrounding the kidney

renal dialysis a process in which technology is used to remove urea and excess water from the body when kidney function is insufficient to do this

renal pelvis an area in the centre of the kidney into which urine from the collecting ducts drains

renal pyramids cone-shaped areas of tissue in the medulla of the kidney, consisting of tubules that transport urine from the outer part of the kidney to the calyces

renal vein vein carrying blood with a low concentration of urea out of the kidney to return it to the heart

Replacement hypothesis (Out of Africa hypothesis, Eve hypothesis) the theory that archaic *Homo sapiens* left Africa and a second migration out of Africa happened about 100 000 years ago, and that modern humans of African origin conquered archaic groups and replaced them by interbreeding with and out-competing them

replication fork the structure formed by the partial separation of the DNA double helix during DNA replication

reproducibility the extent to which the same result can be obtained, within uncertainty, when repeated measurements are made

reproducible giving the same result, within uncertainty, when repeated measurements are made

reproduction the ability of an organism to produce offspring, ensuring the continuity of life

reproductive cloning a type of cloning capable of producing a whole organism that is identical to the donor organism; also known as whole-organism cloning

reproductive success an organism's ability to produce fertile offspring that survive to reproductive maturity and produce offspring of their own, in this way replacing the parent

reproductive technology technology (such as artificial pollination, artificial fertilisation and cloning) used to bring about reproduction or increase the breeding success of an individual

research question the specific question that a particular experiment or investigation is designed to answer

reservoir any substance or tissue that is a source of pathogens that can cause further infection

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response action taken by a tissue or organ to counteract a stimulus and return the body system to the set point

restriction enzyme a type of enzyme made by bacteria that can cut DNA molecules at specific places

retina a thin membrane covering the rear portion of the eye and containing photoreceptors

retinal (retinene) a molecule derived from vitamin A that is one part of a visual pigment

retinitis pigmentosa a disease in which the rods and cone cells in the eye degenerate gradually

retrotransposon inserted viral RNA reverse-transcribed back into DNA

retrovirus virus that contains RNA

rheotaxis a response to a stimulus in which the cell (or organism) swims into an oncoming current

rhizome an underground horizontal modified stem that can split for asexual reproduction

rhodopsin a visual pigment contained in the rod cells of the eye

ribosomal RNA (rRNA) molecule that forms a structural part of ribosomes and is made in the nucleolus of the cell

ribosome a cell organelle responsible for protein synthesis (translation of mRNA into a polypeptide chain)

rickets a disease in children caused by a lack of vitamin D in the diet and resulting in defective calcification of bones, retardation of growth and deformities of the skeleton, such as bowed legs

RNA polymerase an enzyme that attaches to a DNA template strand and begins assembling a complementary strand of RNA, in a process known as transcription

rod a photoreceptor (light-detecting) cell in the retina of the eye

round window a membrane in the middle ear that vibrates in conjunction with the oval window, allowing fluid in the cochlea to move, which then converts the vibration into a nervous signal that goes to the brain

runner a long, thin modified plant stem that grows along the surface of the soil and gives rise to new plantlets by asexual reproduction

S

sampling bias a form of selection bias in which the choice of subjects for a study does not reflect the population being studied

saprophytic describes organisms that live on dead, decaying organic matter

sarcoma a cancer that forms in the connective tissue or muscle, such as bone or blood vessels

scaffold the dense protein centre of the nucleoid of a prokaryotic cell around which the circular DNA of the chromosome is super-coiled.

scatter plot a graphical representation of the relationship between the individual data points of two variables

scientific method a systematic process of observation, experimentation, measurement and analysis to either support or disprove a hypothesis

sclera an opaque, tough protective coat that surrounds the eye

scurvy a disease caused by lack of vitamin C in the diet, resulting in poor wound healing, joint pains, bleeding gums, bones that do not grow or heal and spontaneous haemorrhaging

seasonal breeder a mammal whose reproduction is limited to certain times of the year, during breeding seasons, and who mates only during periods of female fertility

secondary data data or information that has been collected by someone other than you

secondary immune response the response of the immune system when it is exposed to a pathogen on subsequent occasions

secondary sex characteristics features that appear during puberty in humans and at sexual maturity in other animals, as a result of the production of sex hormones, but are not directly part of the reproductive system; examples include deepening of the voice and hairiness in males and development of mammary glands in females.

secondary structure the three-dimensional arrangement of polypeptide chains created by folding or pleating

secondary tumour a tumour that develops from cells that have travelled from a primary tumour to a different part of the body

secretion part of the process of moving toxic substances (e.g. urea, creatinine) from the blood capillaries into the urine; occurs in the renal tubules of the nephron

sector graph *see* pie graph

seed a plant ovule containing an embryo, which is capable of developing into another plant

selection bias a form of bias that results in selection of data or subjects not being random

selective breeding a form of artificial selection by humans deliberately selecting and mating organisms with specific characteristics

sense organ an organ with receptors concentrated in particular areas, such as the eye or the ear

sensorineural hearing loss a form of hearing loss resulting from damage to or malformation of the inner ear, including parts of the cochlea, hair cells or auditory nerve

sensory neuron neuron that carries signals from sensory cells in the peripheral nervous system to the CNS

septicaemia also called blood poisoning; when bacteria or their toxins enter the bloodstream

sequencing finding the exact nucleotide sequence of a certain length of DNA

serotonin a compound in platelets that constricts blood vessels

serotype a group or strain within a species of microorganism, with the same cell surface antigens

set point the ideal or normal value for a specific internal condition

sex chromosomes chromosomes that carry genes that determine the sexual characteristics of a person and influence whether the individual is female or male

sex determination the way in which sex chromosomes separate during meiosis and gamete formation and then recombine during fertilisation to determine whether the offspring will be male or female

sex hormone a hormone that specifically affects the growth or functioning of the reproductive organs or the development of secondary sex characteristics

sex linkage when genes are located on the sex chromosomes and code for characteristics other than the gender of individuals

sex-linked inheritance the inheritance pattern for traits that are located on the genes of the sex chromosomes and therefore may be inherited in different ratios in males and females; appears more frequently in males (linked to the X chromosome) or in males only (linked to the Y chromosome).

sexual reproduction reproduction involving two parents, who produce offspring that contain a mix of the parents' genes and therefore differ from each other and from the parents

short tandem repeats (STR) short sequences of DNA, consisting of 2 to 5 base pairs, that are repeated

silent mutation a mutation involving changes in the DNA sequence that do not cause a change in amino acid and therefore have no effect on the proteins formed

single nucleotide polymorphism (SNP) a variation (change in form) resulting from a single different base (adenine, thymine, cytosine or guanine) present at a particular locus on DNA in the genome of at least one per cent of the individuals in a population.

sister chromatids newly replicated chromatids still joined by a single centromere

SI units International System units of measurement; includes seven base units from which other units are derived: metre (length), kilogram (mass), second (time), kelvin (temperature), ampere (electric current), candela (luminosity) and mole (quantity of a substance)

skin the soft, outer covering of vertebrates

Socratic seminar a group discussion format where students help each other to understand ideas, issues and values

somatic cell a body cell (non-reproductive cell), containing two sets of chromosomes

somatic cell nuclear transfer (SCNT) a type of reproductive cloning that involves three animals: egg donor, nucleus donor and surrogate

somatic mutation change in a DNA sequence that occurs in somatic (non-sexual) cells and may affect only part of an organism

spermatogenesis production of sperm

spontaneous generation the theory that living organisms appear from inorganic matter

spontaneous mutation a natural error arising randomly during DNA replication

sporangia (sing. sporangium) specialised tips of hyphal threads that develop into microscopic spores

spore a unicellular reproductive cell, the product of sexual or asexual reproduction in fungi and some prokaryotes, mosses and ferns; dispersed in large numbers and can develop into a new organism under favourable conditions.

sporozoa any protozoan that can form reproductive cells known as spores, e.g. *Plasmodium* spp.

stem cell an unspecialised cell that is capable of dividing and becoming any type of specialised tissue

sterilise to remove any living microorganisms from a substance or object

sterile microbe-free; uncontaminated

sticky end a section of single-stranded DNA with exposed nucleotide bases at the end of a double-stranded molecule

stigma the sticky tip of the female part of a flower, where pollen is received

stimulus a change in a particular condition in the internal environment of the body; provokes a reaction by a tissue or organ

stirrup (stapes) – the last of the three ossicles in the middle ear; passes vibrations to the oval window

stocking density the number of grazing animals per hectare of land at any one time

structural relating to the physical characteristics of an organism

succulent a plant with adaptations for surviving dry conditions, such as fleshy stems or leaves that can swell and retain moisture

sucker the modified root of a plant, which gives rise to new plants

suppressor T cell T lymphocyte responsible for blocking the action of other lymphocytes, to stop the immune response when the infection has been defeated

surface marker a specific protein on the cell membrane that identifies and characterises the cell as being at a particular stage of specialisation

suspensory ligaments ligaments in the eye that connect the ciliary muscles to the lens

symptom a feature of a disease that is apparent to a patient

synapse a small gap between the axon of one neuron and the dendrite of an adjacent neuron

syndrome a group of symptoms that occur together and characterise a particular disease

systematic error/bias a consistent, repeated error; it may be caused by faulty measuring equipment or bias in the design or conduct of a study

systemic acquired resistance a whole-plant response that occurs following infection with a pathogen

T

target cells cells that possess receptors for a specific hormone that influences the cells' activity

targeted therapies treatments that target specific areas

tectorial membrane the upper membrane of the organ of Corti, against which the hair cells are pushed when a vibrational wave in the perilymph pushes against the basilar membrane

tertiary structure the three-dimensional shape of a protein created by attraction between folded and pleated polypeptides

test cross the breeding of an organism that has a dominant trait with an organism that shows the recessive form of the trait, and analysis of the ratios of the offspring phenotypes, to determine whether the dominant organism is homozygous or heterozygous for that trait.

testosterone an androgen (sex hormone) secreted by cells in the testes; plays a primary role in the production of sperm

tetrad the four haploid daughter cells resulting from the second part of meiosis, meiosis II, where the two haploid cells created during meiosis I each divide again

therapeutic contributing to the healing of a disease

thermoreceptor receptor cell(s) that detects changes in temperatures (stimulus)

thermoregulation regulation of body temperature

thyroid gland a gland consisting of two lobes on either side of the neck, which produces thyroxin

thyroxin an iron-containing hormone responsible for controlling metabolic rate

tolerance limits upper and lower levels of specific conditions in the internal environment of the body

torpor short-term hibernation where the body temperature drops below 30°C, and metabolism, heart rate and respiratory rate decrease, accompanied by a reduced response to external stimuli

trait a particular physical feature or characteristic of an organism

transcription the process of making RNA from a DNA template by the enzyme RNA polymerase

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transcription factor a protein that controls which genes are transcribed

transduction a method of introducing foreign DNA into a cell using a viral vector; the virus is injected into the bloodstream or delivered by aerosol into the human subject

transfer RNA (tRNA) a small RNA molecule twisted into the shape of a clover leaf, each 'leaf' of which attaches to a specific amino acid at one end and has a triplet of bases (an anticodon) at the other end to recognise and pair with mRNA; a vehicle of amino acid transport in protein synthesis

transgenic species a species whose genome includes DNA from another species

translation the synthesis of a polypeptide sequence from mRNA, with the aid of tRNA, on a ribosome

translocation a chromosomal abnormality resulting when a section of one chromosome joins to a chromosome in a different homologous pair

transpiration the evaporation of water in a plant through the stomata in a leaf

transposon transposed DNA (also known as 'jumping genes' or transposable elements); a DNA sequence that spontaneously moves from one location on the genome to another and disrupts DNA functioning in its new location on the chromosome

treatment administration of remedies (pharmaceutical, behavioural, surgical) to an individual suffering from a disease

trisomy a genetic condition in which the total number of chromosomes present is one more than the normal diploid number of chromosomes

tumour suppressor gene gene that codes for proteins that slow down or stop cell growth and mitosis

tympanic membrane the eardrum; vibrates as sound waves enter through the auditory canal and transmits the vibrations to the middle ear

U

ultraviolet radiation that part of the electromagnetic spectrum where wavelengths are shorter than those of visible light, making it damaging to DNA

uncertainty an estimate of the range of values within which the 'true value' of a measurement or derived quantity lies

ureter tube that transfers urine from the kidney to the bladder

urethra tube that transfers urine from the bladder to exit the body

urine liquid containing water and waste material filtered from the blood in the kidneys

V

vaccination administration of a vaccine

vaccine a suspension that contains an attenuated or killed pathogen or toxin that causes an immune response so that immunity is conferred to the organism receiving the vaccine

valid relating to results that are affected by a single independent variable and hence any differences are due to that variable

variability the different forms of a gene within a population; the total of all alleles present in the gene pool of a population

variation the differences in characteristics evident in individuals

vasoconstriction the process by which the blood vessels constrict (narrow), removing blood from the skin surface to conserve heat

vasodilation the process by which blood vessels dilate (expand), bringing blood closer to the skin and allowing heat to escape

vector an insect that transmits pathogens by biting; an example is an *Anopheles* mosquito transmitting *Plasmodium*, which causes malaria

vegetative propagation a type of asexual reproduction in plants, in which new individuals arise from portions of the root, stem, leaves or buds of adult individuals and are genetically identical to the parent

vertical transmission viral spread from parent to offspring

virulence factor molecule produced by a pathogen that increases its effectiveness in invading the host and causing disease

visual acuity the ability to see a clear and precise image

vitreous humour a jelly-like, clear fluid between the lens and the retina; refracts light and helps to maintain the shape of the eyeball

viviparous describes animals in which the embryo develops inside the female parent's body, obtaining nutrients through a placenta and is subsequently born alive

volunteer bias a form of selection bias in which volunteers who participate in the study have a vested interest in the condition being studied

W

whole-organism cloning the production of an exact copy of a whole organism; also called reproductive cloning

X

xenotransplantation use of organs from other animals for transplantation into humans

xerophyte plant that lives in arid conditions and has adaptations to balance the opening of stomata for gas exchange with conservation of water

Z

zoonosis an infectious disease that can be transmitted from animals to humans

zygote fertilised egg

INDEX

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ISBN 978-0170485067