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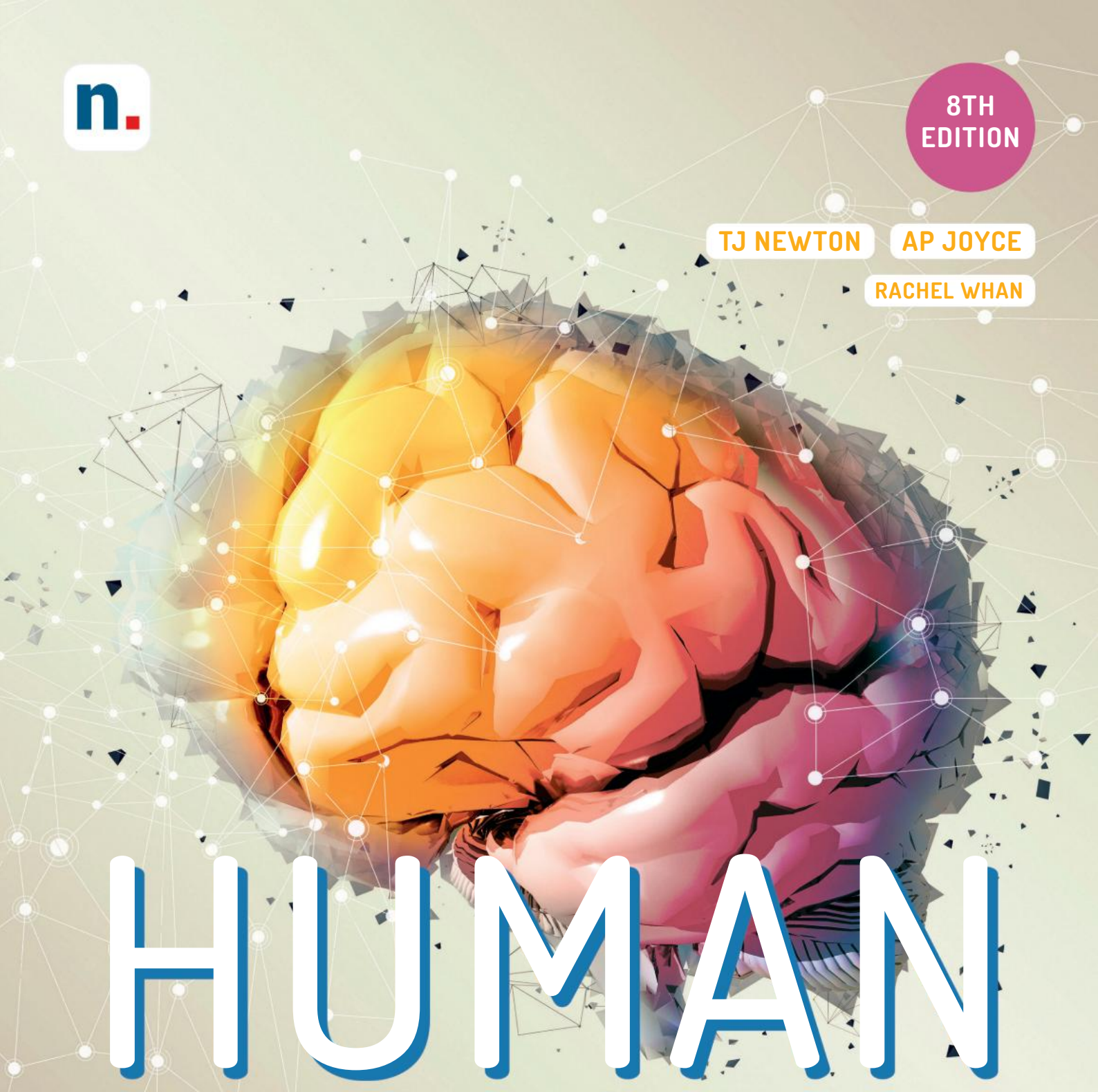

HUMAN PERSPECTIVES

ATAR UNITS 3 & 4

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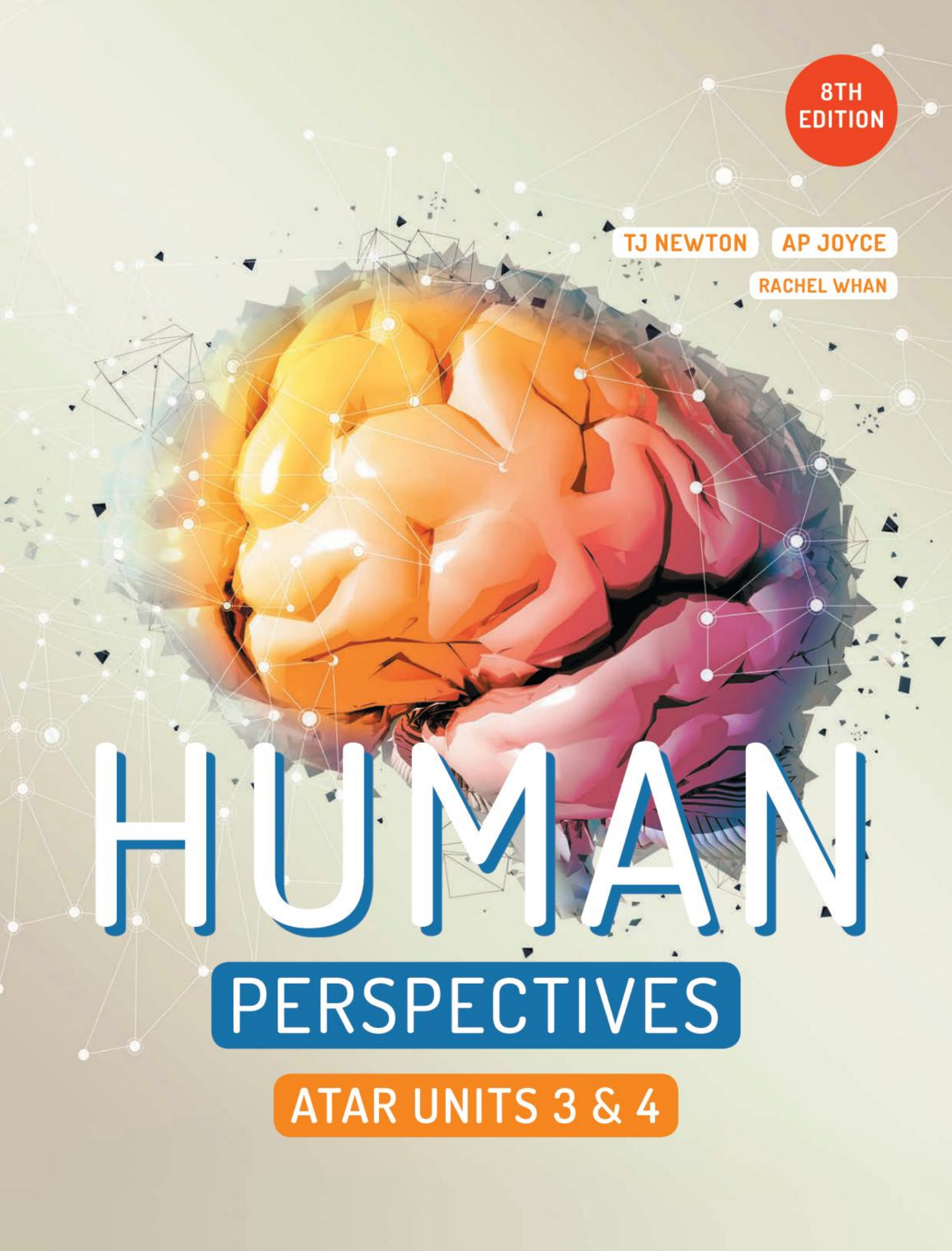

HUMAN

PERSPECTIVES

ATAR UNITS 3 & 4

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Human Perspectives ATAR Units 3 & 4
8th Edition
TJ Newton
AP Joyce
Rachel Whan
ISBN 9780170449168

Publisher: Sarah Craig
Project editor: Robyn Beaver
Copy editor: Robyn Flemming
Text design: Rina Gargano (Alba Design)
Cover design: Jenna Lee Fai (Jenki)
Project designer: Justin Lim
Cover image: Getty Images/Victor Habbick Visions/Science Photo Library
Permissions researcher: Helen Mammides
Production controller: Alice Kane
Typeset by: SPi Global

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.

Acknowledgements
Extracts from the Human Biology ATAR course Year 12 syllabus are used by permission, School Curriculum and Standards Authority. 2020 Human Biology ATAR Year 12 course syllabus: Homeostasis and disease unit content and 2020 Human Biology ATAR Year 12 course syllabus: Human variation and evolution unit content. The School Curriculum and Standards Authority does not endorse this publication or product.

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 7, 80 Dorcas Street
South Melbourne, Victoria Australia 3205

Cengage Learning New Zealand
Unit 4B Rosedale Office Park
331 Rosedale Road, Albany, North Shore 0632, NZ

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Printed in China by 1010 Printing International Limited.
1 2 3 4 5 6 7 24 23 22 21 20

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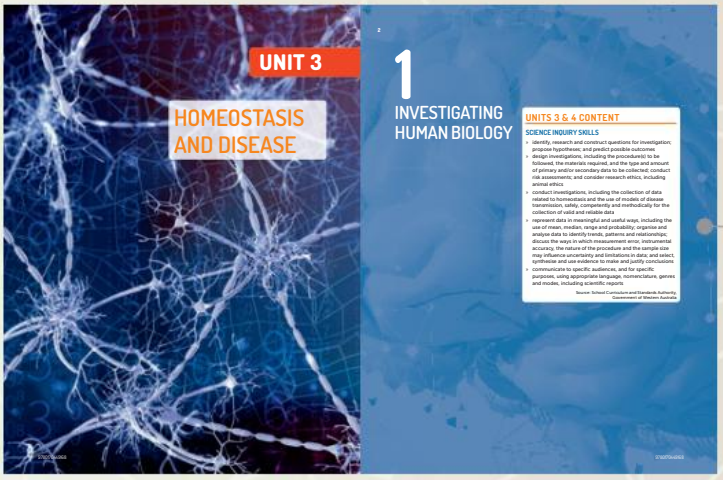
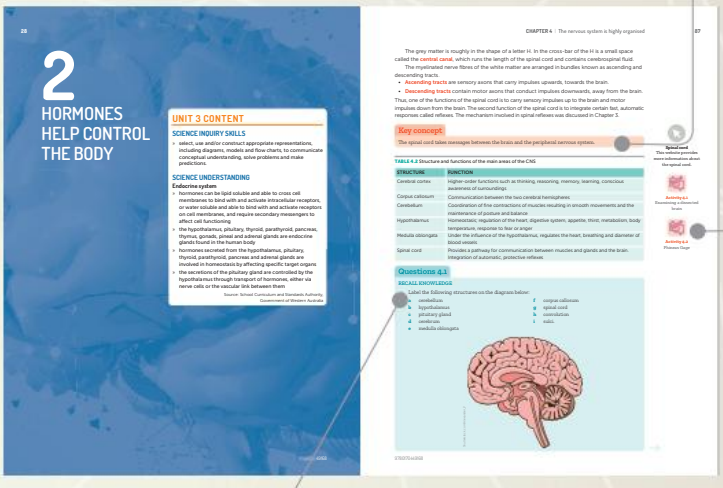
USING *HUMAN PERSPECTIVES*

Human Perspectives has been comprehensively updated to address all aspects of the School Curriculum and Standards Authority (SCSA) Human Biology ATAR course. This series will enable you, the student, to achieve maximum understanding and success in this subject. Each page has been carefully considered to provide all the information you need in a variety of different formats such as text, figures and links to online material. You will find it easy to navigate through each chapter and see connections to the practical activities and investigations through the use of icons, highlighting the importance of the interconnectedness between the conceptual and practical aspects of Human Biology.

Each chapter begins with a **chapter opening page**, which presents the learning outcomes to be covered in the chapter under the Science Inquiry Skills, Science as a Human Endeavour and Science Understanding strands from the SCSA Human Biology ATAR syllabus.



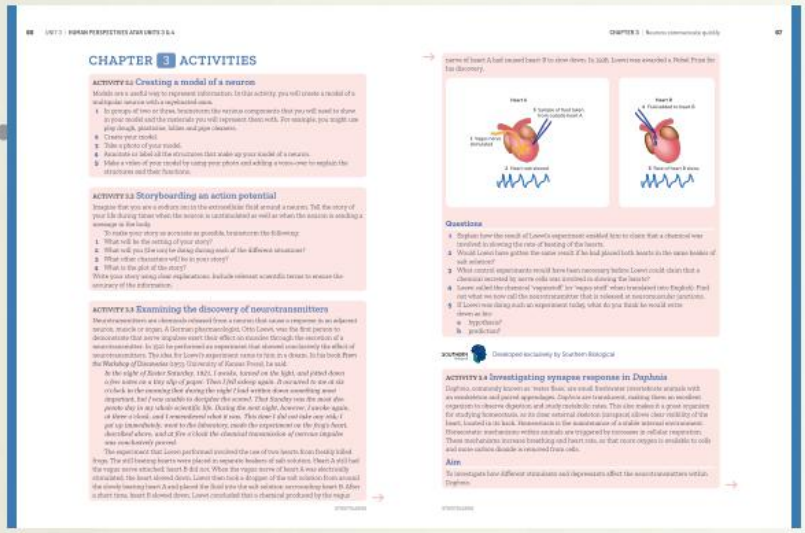
Important ideas, concepts and theories are summarised in **key concept boxes** throughout the chapters. These provide reinforcement and summary for improved assimilation of new ideas.



Regular opportunities to recall new terms and facts, and to apply concepts, are provided in **question sets** at the end of each chapter section.

Connections to practical activities and investigations are indicated using **margin icons**. Interactive icons link to digital worksheets and websites.

Practical activities and investigations introduce and reinforce the Science Inquiry Skills listed in the SCSA Human Biology ATAR syllabus. They allow you to discover and explore the living world.



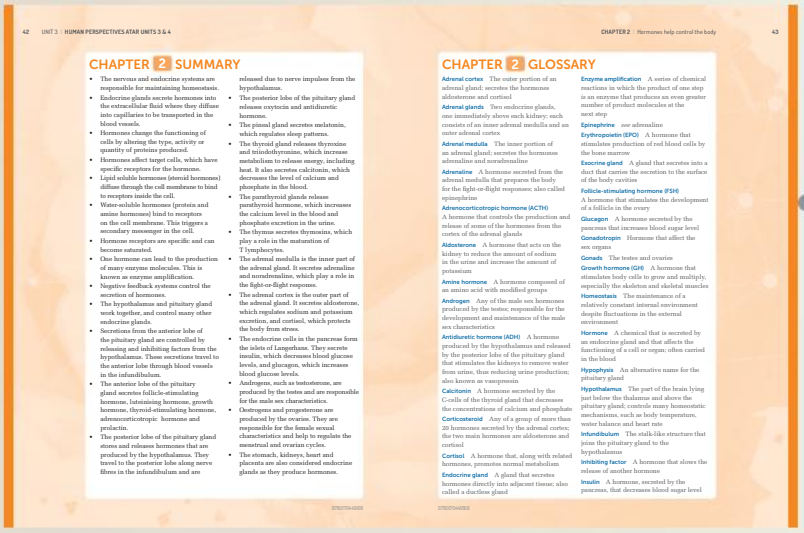
CHAPTER 3 ACTIVITIES

ACTIVITY 3.1 Creating a model of a neuron

ACTIVITY 3.2 Storyboarding an action potential

ACTIVITY 3.3 Examining the discovery of neurotransmitters

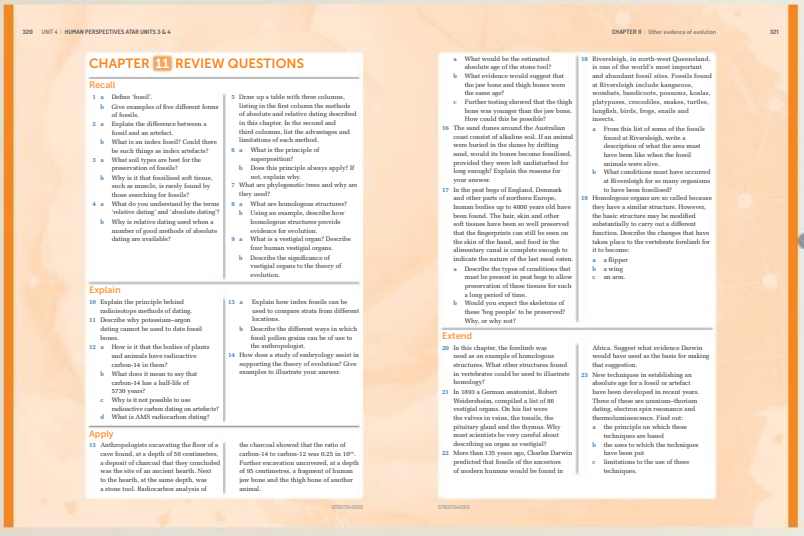
ACTIVITY 3.4 Investigating synapse response in Daphnia



CHAPTER 2 SUMMARY

CHAPTER 2 GLOSSARY

Chapter review sections include a **chapter summary** for revision and reflection, a **glossary** of important terms introduced in the chapter, and **review questions** that check understanding and provide opportunities for application and analysis of concepts.



CHAPTER 11 REVIEW QUESTIONS

Recall

Explain

Apply

Extend

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Teachers will have access to these resources plus answers to all student book questions and activities, practice tests, chapter PowerPoints, and syllabus mapping and teaching plan documents. Investigation support has been provided by Southern Biological and includes lab advice and safety sheets, and videos to assist both the teacher and laboratory technician in preparing and delivering the investigation to students.

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AUTHOR ACKNOWLEDGEMENTS

A great many people have contributed to the writing and production of this book; and to all those people, the authors express their sincere thanks.

For previous editions, we are particularly indebted to June Gouldthorp and Pauline Charman. We wish to acknowledge again their vital contributions. June provided invaluable feedback on the original draft of the manuscript and helped ensure all content areas had been covered. Pauline, then Project Officer for Human Biological Sciences at the Curriculum Council of Western Australia, offered clarification of the content. We also remain grateful for the work of Audrey Sewell-Smith, Jill Lamble and Anabel Kanakis, who reviewed parts of that manuscript and provided constructive criticisms and suggestions.

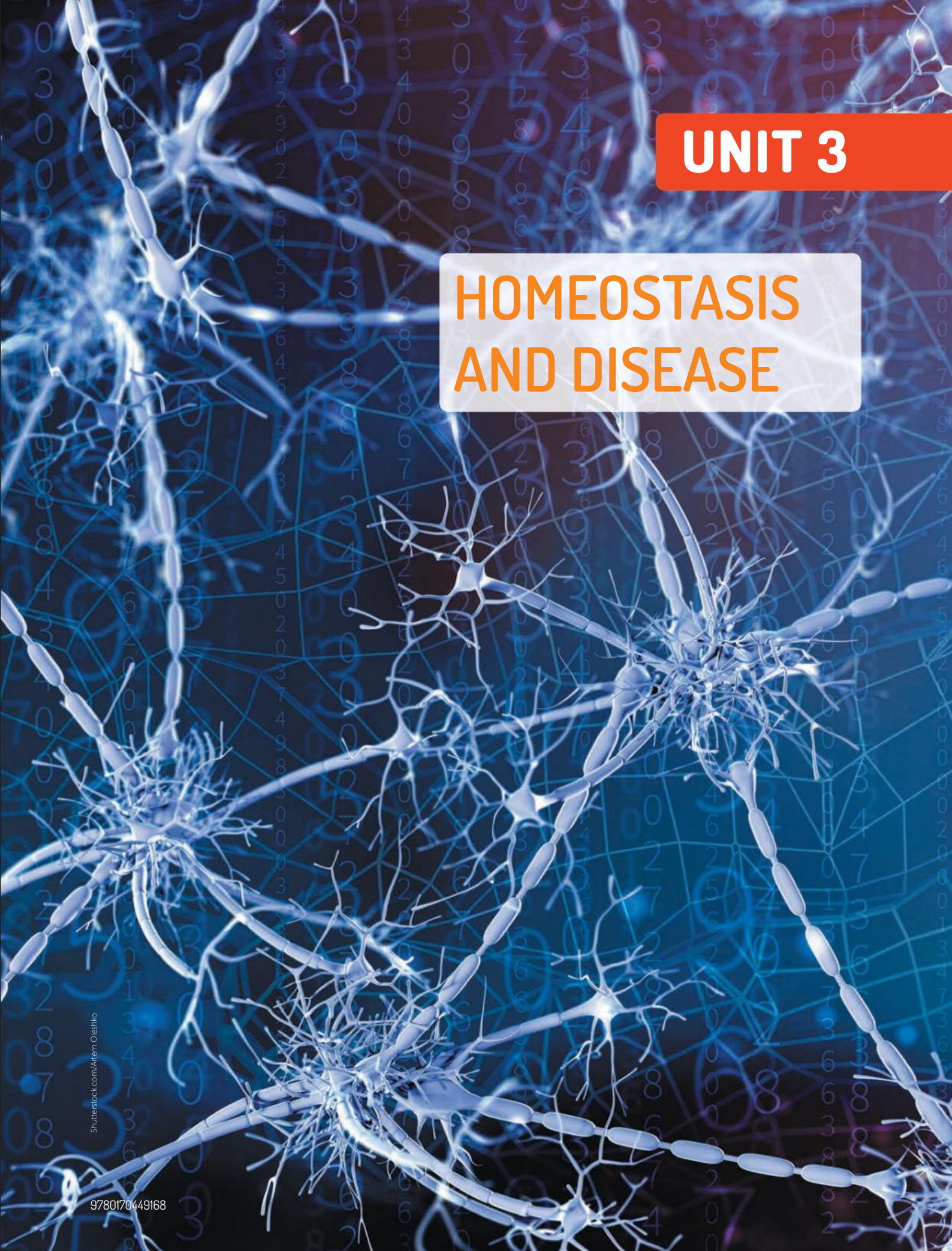
We would also like to thank those people who provided information, photographs, or both, for earlier editions. A special expression of thanks must go to Geoff Meyer of the Centre for Human Biology, University of Western Australia, for his particular contribution to past editions.

For this edition, we extend our gratitude to Derek Cumpsty for writing the practice tests on Cognero Assess, Charlotte Donovan for her valuable feedback on the manuscript and contributions to the digital resources, and Michelle Moreton for her review of the manuscript.

Terry Newton
Ashley Joyce
Rachel Whan

UNIT 3

HOMEOSTASIS AND DISEASE



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1 INVESTIGATING HUMAN BIOLOGY

UNITS 3 & 4 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including the collection of data related to homeostasis and the use of models of disease transmission, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

Source: School Curriculum and Standards Authority, Government of Western Australia

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Herbert Spencer, an English philosopher who lived from 1820 to 1903, neatly summarised what science is all about: science is a process of inquiry aimed at finding answers to problems and discovering new knowledge about the natural world. The knowledge discovered as a result of scientific inquiry becomes a part of science. That is, science means two things: a process of discovery and the knowledge that is discovered. The information presented in this book is science. It is some of our present knowledge about the human species, knowledge that has been obtained by scientific investigation.

1.1 TYPES OF INVESTIGATIONS

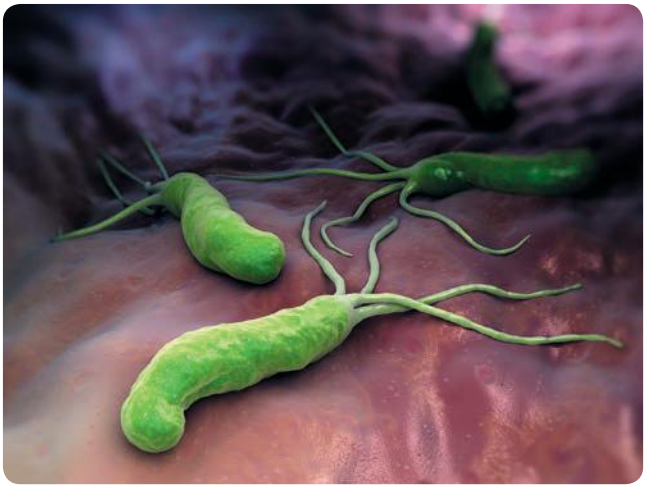
Scientists use a range of techniques to expand our knowledge.

Observations

Observation is an essential part of science. Any investigation, regardless of the procedure used, will involve some form of observation. In investigations based on observation, scientists are looking for patterns. When a pattern becomes evident it may be possible to draw tentative conclusions.

An example of an investigation based on observation is the discovery that peptic ulcers are caused by a bacterial infection. In 1979, Dr Robin Warren, a pathologist at the Royal Perth Hospital, observed the presence of bacteria in samples of stomach tissue that he was examining. Continued observation over the next few years showed that the bacteria were often present in the stomachs of patients suffering from stomach inflammation. Warren's discovery was not taken seriously because, at the time, it was believed that the stomach contents were too acidic for bacteria to survive.

In collaboration with Dr Barry Marshall, a doctor specialising in stomach disease, Warren was able to show that a particular species of bacterium was present in the majority of cases of stomach and duodenal ulcers. They also found that it was rare to have an ulcer without being infected by the bacterium that Warren had discovered. Marshall and Warren went on to culture the bacterium and to show that it did indeed cause ulcers. As a result of their discovery it became easy to treat stomach and duodenal ulcers with antibiotics. The discovery that the bacterium *Helicobacter pylori* was the cause of ulcers began with simple observation and became one of the most significant events in Australian medical history. Robin Warren and Barry Marshall were awarded the Nobel Prize in Physiology or Medicine in 2005.



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What is *Helicobacter pylori?*

FIGURE 1.1 *Helicobacter pylori* bacteria

Another example of systematic observation is when it is used to gain knowledge of animal behaviour. Jane Goodall, the first person to observe the social organisation of chimpanzees in the wild, documented the interactions of chimps with one another, their social hierarchy, their tool making and many other features of their society. She began her observations in 1960, and her work is being continued today through the Jane Goodall Institute.

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FIGURE 1.2 Jane Goodall observing the behaviour of chimpanzees

Controlled experiments

Controlled experiments, sometimes called fair tests, are designed to investigate relationships between factors (or variables). They involve changing one variable while all the other variables are kept the same. Any differences in the results should be due to the changed variable.

Howard Florey, an Australian working at Oxford in England, used controlled experiments to demonstrate the effectiveness of penicillin in treating bacterial infections. In 1940 a crucial experiment to test the effectiveness of penicillin as an antibiotic was carried out. Eight mice, all the same weight and age, were each injected with 100 million streptococci, a type of bacterium. Previous experiments had shown that an injection of that size would kill all mice injected. After the injection of streptococci, four of the mice were put back in their cages and given no further treatment. The other four mice were given injections of penicillin. The mice in the control group – those that did not receive penicillin – all died within 12 hours. Mice in the experimental group, which were given penicillin, survived for many days – one for more than six weeks.

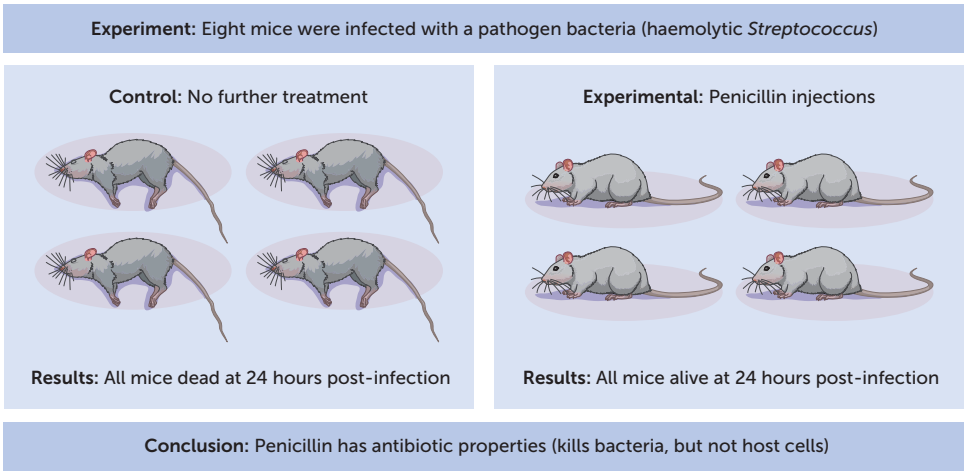


FIGURE 1.3 A controlled experiment was used to investigate the effectiveness of penicillin

The only difference between the mice in the two groups was the injection of penicillin, so the survival of those mice was good evidence that penicillin was effective in combating bacterial infections. Further positive feedback from repeated controlled experiments gave Florey and his co-workers the confidence to try penicillin on humans suffering from bacterial infections. The results were outstanding. Florey and his colleague Ernst Chain were awarded the Nobel Prize in Physiology or Medicine in 1945.

Surveys

A **survey** is a process of systematically collecting, analysing and interpreting information about an aspect of a study. Surveys are usually designed to collect data from a large number of subjects. The information may be collected using a questionnaire or by interview. Using the large amount of information collected, the researcher can then look for patterns in the data.

Dr Karl's survey

Dr Karl Kruszelnicki was stimulated by a listener to his radio program to carry out a survey into the origins of belly button lint. The survey was conducted over the Internet, with 4799 people responding, and publicised on the radio station Triple J. The patterns in the responses revealed that people more likely to have belly button lint are male, hairy, with a concave belly button, and that the amount of lint increases with age. This was a light-hearted exercise, but it does demonstrate the principles involved in conducting a survey. In 2002, 'Dr Karl' received an Ig Nobel Prize for his efforts. Ig Nobel Prizes are awarded for research that makes people laugh and then makes them think.

Trial and error

Trial and error sounds like a random process, but when used in scientific research it is systematic. The process involves one attempt to solve a problem being followed by another. Each trial is recorded, and the results allow the investigator to gradually home in on the solution to a problem. Thomas Edison, who developed the electric light globe, had to find a suitable material for the filament in the light globe. Using trial and error, he examined more than 600 different materials before finding one that was satisfactory.

Many new drugs, such as antibiotics, have been discovered using trial and error. Chemical compounds extracted from plants can be tested on cultures of bacteria to see whether they have any effect. Those that show promise can then be subjected to further testing under different conditions. Meticulous records of the results of each trial must be kept. Such research is often prolonged and tedious, but it is often the only way to find effective substances.

Case studies

A **case study** is an in-depth investigation of one particular person or situation. Case studies are frequently used in areas such as education and business management. However, they may also be useful in some areas of science. For example, in medicine the progress of a particular disease in one person may be documented. Such a case study can extend or help to confirm what is already known about the disease.

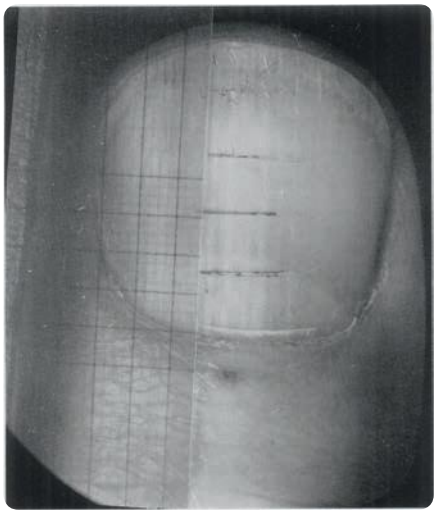
Longitudinal studies

A **longitudinal study** is conducted over a long period of time. It is similar to a case study but is more prolonged. Longitudinal studies may take place over many years, even decades. They can also be done retrospectively, which means that the researcher can examine records of past events to build up a picture of change over time.

Busselton Health Study

The Busselton Health Study is a longitudinal study of the population of Busselton, a coastal town in the south-west of Western Australia. Begun in 1966 and continuing today, it is the world's longest-running study of the health of a population.

A longitudinal study on a smaller scale was carried out by an American doctor, William Bean, who studied the growth of his fingernails for 35 years. He did this by filing a horizontal line on his thumbnail just above the cuticle and recording how long it took the mark to reach the tip of his thumbnail. From his records he was able to calculate the growth rate. In 1980, after 35 years of measurements, Bean was able to conclude that the growth of his nails had slowed from 0.123 mm a day when he was 32 years of age to 0.095 mm a day at the age of 67.



William B Bean, 'A Discourse on Nail Growth and Unusual Fingernails' American Clinical and Climatological Association (1962; 74: 152-167)

FIGURE 1.4 Dr William Bean used lines in his fingernails to study their growth rate in a longitudinal study

Key concept

Scientists use a range of different methods to acquire new knowledge, including observations, controlled experiments, surveys, trial and error, case studies and longitudinal studies.

Questions 1.1

RECALL KNOWLEDGE

1. List the methods of investigation used in science.
2. Describe how observation led to our current understanding of stomach and duodenal ulcers in humans.
3. Classify each of the following as either a controlled experiment, survey, case study or longitudinal study.
   - a. A scientist analysed information about the diets and health conditions of 500 people in Western Australia.
   - b. The ability to concentrate in school was investigated in 30 students. Fifteen students drank only water at breakfast, while the other 15 students drank coffee with the same breakfast.
   - c. Casey suffers from epilepsy. Her doctors have been keeping records of the frequency, length and triggers for her seizures for the last 30 years.

APPLY KNOWLEDGE

4. Discuss the implications for an investigation if more than one variable is changed.
5. Explain why trial and error is often a long investigation process.
6. State how case studies and longitudinal studies are:
   - a. similar
   - b. different.

1.2 CONDUCTING INVESTIGATIONS

As we have seen, there are many ways of conducting investigations in science; however, they are all done in a methodical and systematic way. The exact method used will be the one that best suits the situation. Many investigations lead to the testing of a hypothesis and follow a similar pattern, called the **scientific method**, as follows:

1. Recognise a problem and define a question.
2. Collect as much information as possible relating to the problem.
3. Propose a **hypothesis** – a possible explanation for the problem. The hypothesis is written as a statement that can be tested.

4 Test the hypothesis using an experiment.
5 Analyse and interpret the data collected from the experiment.
6 Draw conclusions about whether the hypothesis was supported or disproved.
7 Report on the investigation.

Note that although a hypothesis may be disproved, it cannot be proved. The results of an experiment can only provide *support* for the hypothesis. As Albert Einstein said:

> *No amount of experimentation can ever prove me right; a single experiment can prove me wrong.*

The scientific method outlined above can be applied to many problem-solving situations. Figure 1.5 shows how a doctor uses the scientific method to arrive at a diagnosis. A mechanic would probably use the same method to solve the problem of why a car won't start.

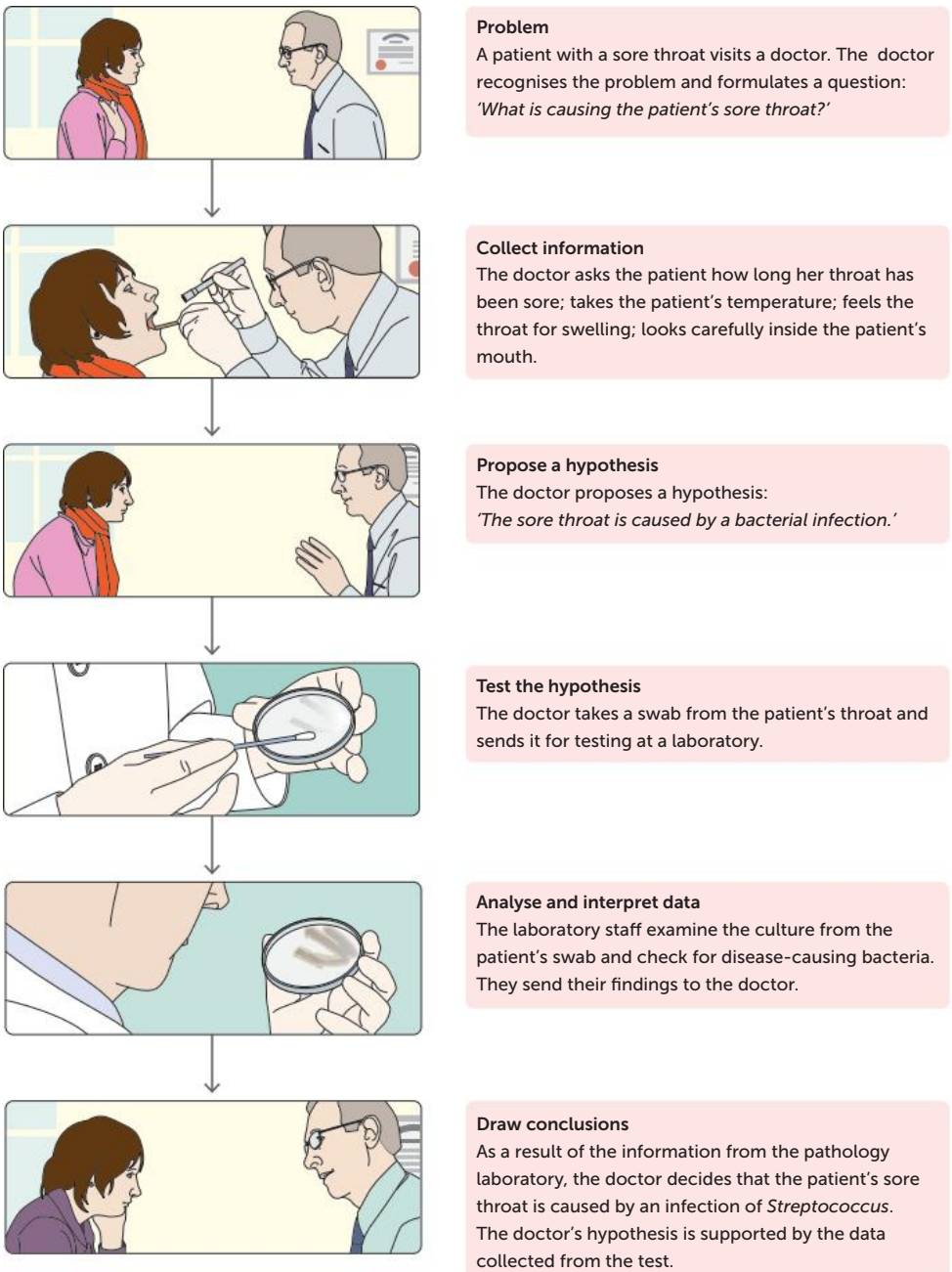


FIGURE 1.5
The scientific method

Scientific method
This website has one interactive that describes the scientific method and another that shows how it is applied in astronomy.

How science works
This website provides a series of modules designed to answer the question: 'What is science and how does it work?'

Key concept

The scientific method provides a systematic approach to conducting investigations in order to test a hypothesis.

Some scientists are critical of descriptions of the 'scientific method' because of the many different ways of gathering data. One of the main criticisms of using this 'method' as a model of scientific investigation is that it does not give enough weight to the importance of observation as a means of obtaining knowledge. In some investigations, observation alone can lead to new understanding without the need for any experimentation.

Despite the criticisms, the scientific method is a useful model because it can be applied to many investigations. When you conduct investigations yourself, you will find the model a useful planning tool.

Literature review

One of the early steps in the scientific method is to 'collect as much information relating to the problem as possible'. One way of collecting information is to carry out a literature review. A **literature review** is a survey of the material that has been written about the subject under consideration. Literature reviews used to involve long hours of library research; however, the Internet has made them much easier to carry out.

The purposes of a literature review are as follows:

- To help the researcher define the problem: defining the problem carefully helps in the design of an investigation that will contribute to solving the problem.
- To find out what is already known about the problem: this prevents duplication of effort and allows the researcher to build on knowledge that is already available.
- To assess research methods: methods used by others may be used or adapted for the researcher's own investigation.
- To allow researchers to relate their findings to what is already known: this is particularly useful for the final report on an investigation. It is also helpful in considering areas for further research.

Safety

It is important that an investigation presents no danger to the participants or to the investigators. Examine the design of your investigation to make sure that any associated safety risks are minimised and controlled. Consider the need for safety devices such as fire extinguishers, fire blankets, fume hoods and eye washes. Also assess the need for personal protection such as safety glasses, gloves, face masks and protective clothing. For some investigations it may be necessary to assess the participants. For example, do they have any allergies to the substances being used? Do they have health problems that could be affected by the activities involved?

Safety considerations should include not just the physical safety of the people involved, but also factors such as whether the participants will feel insecure, threatened or embarrassed.

Ethics

Ethics are a set of moral principles or values. They are standards that are observed by most people in our society. **Ethical behaviour** is behaviour that follows those principles or values. In scientific research, especially research involving human participants, there are many ethical issues.

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The following are some of the principles that an investigation involving humans must satisfy if it is to be ethically sound:

- *Voluntary participation* – the subjects should not be pressured into taking part in the investigation.
- *Informed consent* – the subjects should be fully informed about the objectives of the research, the procedures to be followed, any possible risks and the potential benefits of the research; consent should only be sought after all information has been given.
- *No risk of harm* – as mentioned in the section on safety, there should be no risk of physical or psychological harm.
- *Confidentiality* – the identities of participants will not be revealed except to people directly involved in the study.

Just as humans must be treated in an ethical way, so too must animals. The requirements for investigations involving animals are set out in the *Australian Code for the Care and Use of Animals for Scientific Purposes, 8th edition (2013)*. The code sets out detailed requirements, but in general terms any use of animals in research or teaching should be:

- valid
- humane
- justifiable
- considerate.

Controlling variables

A **variable** is any factor that may change during an experiment.

The **independent variable** is the factor that is being investigated – it is deliberately changed to determine its effect. This variable is deliberately made different in the control and the experimental groups in an experiment. By comparing the results from the control and experimental groups, the effect of the independent variable can be determined.

The independent variable may also be called the experimental variable or the manipulated variable.

The **dependent variable** is the factor that changes in response to the changes made to the independent variable. It is sometimes called the responding variable. The hypothesis should state the predicted relationship, or trend, between the independent variable and the dependent variable.

Controlled variables are the factors that are kept the same for both the control and the experimental groups in an experiment.

In any experiment it is important that, with the exception of the independent variable, all variables are kept the same for the control and the experimental group of subjects. If one or more is not kept the same, it is impossible to tell which variable is causing any difference between the two groups of subjects.

Sometimes it is difficult or impossible to keep all variables the same. **Uncontrolled variables** are variables that are not kept the same for the control and the experimental groups in an experiment. They may have been overlooked by the experimenter, or they may have been impossible to control. If there are uncontrolled variables in an investigation, this must be taken into account when interpreting the results.

Repetition and replication

Scientific experiments always involve repetition or replication. **Repetition** means doing the same experiment many times. **Replication** means having a number of identical experiments running together or performing the experiment on a large number of subjects at the same time. Both repetition and replication help to demonstrate that results are consistent. If results are different each time an experiment is performed, they are of little value.

Repetition and replication can also help to overcome the effects of uncontrolled variables. For example, if 10 subjects are used in an experiment and one of them is unusual in some way, it will have a big effect on the overall result. If 100 subjects were used, one unusual subject in 100 would not have much effect on the average result.

In designing any experiment, plan for as much repetition or replication as time and resources will allow.

Validity, accuracy and reliability of results

When an experiment tests what it is supposed to test, it has **validity**. Some scientists were testing the hypothesis that 'consumption of junk food affects people's memory'. They fed one group of young rats on fatty food for 12 weeks, and fed another group of older rats a low-fat diet. The rats' memories were then tested using an activity that involved pressing a lever. The rats fed on junk food were more forgetful, so it was concluded that the hypothesis was supported. This experiment did not test what it was supposed to test, for two reasons.

- Testing one species, rats, will only demonstrate the effect on the memory of rats, not any other species.
- Rats' memories may be affected by age. The two groups of rats should have been of the same age.

Experiments can also be invalid if there are uncontrolled variables – that is, if there are factors that could affect the result of an experiment that are not kept the same for the experimental and the control set-ups. When experimenting with humans, it is often very difficult to design a valid experiment because it is hard to control all the variables.

iStock.com/PeopleImages

FIGURE 1.6 Where possible, design an experiment so that the results are expressed as measurements. Measuring height in millimetres is much more meaningful than observations such as 'tall' or 'short'

The **accuracy** of data refers to how close the data is to the exact value. Accuracy is dependent on the equipment used, which needs to be calibrated correctly. For example, if an investigation involved measuring the mass is eaten, it would be more accurate to weigh the food with a laboratory balance than with bathroom scales. The balance would also need to be zeroed prior to weighing the food, for the mass to be accurate.

Reliability is the extent to which an experiment gives the same result each time it is performed. The measuring instruments used in the experiment must also be reliable; that is, they must give the same measurement each time they are used. For example, you may have a set of bathroom scales that give three different weights when you step on them three separate times. Those scales are unreliable, and if used in an experiment would make the results unreliable. The bathroom scales may give the same reading every time but it may be consistently higher or lower than the actual weight. In that case, the scales are reliable, but inaccurate.

Repetition and replication are used to identify that results are reliable, but they do not improve the accuracy of the experiment.

Key concept

During a controlled experiment the independent variable is changed to determine its effect on the dependent variable. All other variables are controlled so that the investigation is valid. The correct equipment is chosen and calibrated to ensure accuracy, and the experiment is repeated and replicated to ensure the data is reliable.

Types of data

Data from an investigation can be one of two types:

- **quantitative data** – expressed in numbers and usually involving measurement; for example, 'the students are 174 and 176 cm in height'
- **qualitative data** – observations that do not involve numbers or measurement; for example, 'student A is taller than student B'.

Wherever possible, you should design an investigation so that the results are quantifiable. Numerical results can be ranked, averaged and manipulated in other ways. They can also be summarised using graphs.

Sometimes it is possible to quantify qualitative data. For example, if asking people's opinions on something, they can be asked 'Do you disagree strongly, disagree, agree or agree strongly?' or asked to answer using a numerical value such as 1 for 'disagree strongly' to 4 for 'agree strongly'.

Secondary data

Secondary data is data that has been collected by someone other than the people who are using the data. For example, earlier in this chapter we quoted the rate of growth of Dr William Bean's fingernails. This is secondary data – fingernail growth was measured by Dr Bean, not by the authors of this book.

Errors and limitations in data

It is important that data is checked carefully for errors. In science, an error is not necessarily a mistake. Rather, it is any deviation from the result that should have been obtained. One of the reasons why scientists provide comprehensive reports on their investigations is so that others can check their data for errors.

Measurements made with any measuring instrument are approximate. For example, if you measure a person's height at several different times, the measurements are unlikely to be the same every time. This may be because there is natural variation in the subject, variation in the measurement process, or both. This uncertainty in measurement is called **measurement error**. In this case the word 'error' does not mean the same as 'mistake'. Your measurements are not wrong; the measurement error is the difference between the measurements you made and the true value of what you were measuring. Repetition can help to reduce measurement error, but it cannot overcome error caused by the limitations or deficiencies of the measuring instrument.

It is also important to understand the limitations of data obtained from an investigation. You must not draw conclusions that go beyond the data. Sometimes it is difficult to look objectively at data, and even experienced scientists can draw conclusions that are not necessarily supported by their data.

One example of reading too much into the data obtained in an investigation arose from a report by Norwegian scientists on the incidence of breast cancer in 25 624 women. Published in the *New England Journal of Medicine* (1997, vol. 336, no. 18, p. 1269), a prestigious medical journal, the results of the scientists' survey showed that the incidence of breast cancer in women who exercise regularly

was reduced by 37%. The media reported on the investigation with headlines stating that 'exercise prevents breast cancer'.

Other scientists pointed out that women who exercise regularly are also likely to be non-smokers, drink less alcohol, have healthier diets, and have higher levels of education and higher incomes than women who do not exercise. Which of these variables was actually contributing to the reduction in breast cancer? Was it really exercise, or could it be having a healthier diet, being a non-smoker, having a better education, and so on? Could it be a combination of some of these factors? Each factor and combination of factors would have to be investigated before arriving at a firm conclusion. This example illustrates some of the pitfalls in analysing data.

Data may sometimes include a *confidence interval*. A confidence interval is used to indicate the reliability of data. It is the range of values above and below a result in which the actual value is likely to fall. For example, opinion polls published in the media may say that 53% plus or minus 1.5% of people will vote for Party X. The confidence interval is 51.5% to 54.5%. A confidence *interval* should be quoted along with a confidence *level*. The *confidence level* most commonly used in research is 95%. This means that if the research were repeated a number of times, the range of values obtained would contain the true value 95% of the time. In the survey of voters, if the survey were repeated many times, the proportion of people who intend to vote for Party X would be between 51.5% and 54.5%, 95% of the time.

Another example may help to clarify this concept. Suppose you wanted to find the average height of Year 12 students in Western Australia. You could measure the height of every Year 12 student in the state and then calculate the average height. This would give you an accurate result (the true value), but it would be impractical. A more practical method would be to measure a sample of Year 12 students. If you took a sample of 20 Year 12 students and calculated their average height it would give you an estimated result, but it would not tell you how certain you could be that your result was correct. Using a mathematical formula, a confidence interval could be calculated that would indicate the reliability of your estimate. The calculated confidence interval may show, for example, that using the same sampling method, the average height of Year 12 students will be between 167 and 179 cm, 95% of the time.

Processing data

If you have designed an experiment to give quantitative data, you will end up with a mass of figures that you must interpret. In a controlled experiment you will have to compare the control and the experimental results. There are some simple calculations that you can do to make the numbers more meaningful.

Average

In science a description of a set of numbers almost always includes a measure of its centre, or its **average**. Averages are a very common and simple way of handling sets of numerical data. The average that is most often calculated is the **arithmetic mean**, often just called the **mean**. To calculate the mean of a group of measurements, you add up all the measurements in the group and divide by the total number of measurements.

Sometimes in a set of measurements there will be values that are well beyond the range of the rest of the measurements. These are called **outliers**. The mean is affected by outliers, because a very high or very low outlier value will make the mean higher or lower than it would be without the outlier included. Outliers may result from mistakes in measurement, the failure of equipment or other errors. If the outliers clearly result from an error, they may be excluded when the mean is calculated.

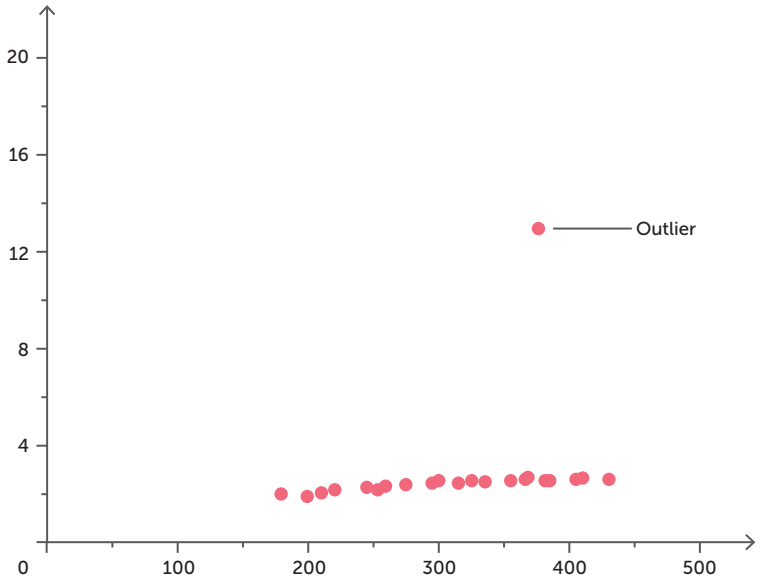

FIGURE 1.7
An outlier is beyond the range of the rest of the data

Median

The **median** is the middle of a set of numbers. It divides the lower set of numbers from the upper set. For example, the heights of the members of a cricket team were measured and (in centimetres) they were: 164, 176, 177, 177, 178, 181, 182, 182, 183, 185, 191.

The median height of the team was 181 cm; that is, 181 is the middle value – there are five team members with heights lower than 181 cm and five with heights higher. If there is an even number of measurements, then the median is taken as the mean of the two values in the middle of the set of numbers.

Using the median of a set of numbers reduces the influence of outliers. Outliers due to measurement error could have a significant effect on the mean of a set of numbers, but would have much less effect on the median.

Range

A measure of the centre of a group of numbers can be misleading. The mean, or the median, gives us no idea about whether all the values are clustered around the centre or whether there is a very wide spread from highest to lowest value. Any description of a set of numbers should therefore include both a measure of centre and a measure of spread.

The simplest way to indicate the spread is to quote the **range** – that is, the highest and lowest measurements in the group. For example, we could say that the heights of students in a Year 12 class ranged from 151 to 183 cm, with a mean of 171 cm.

Ratios and rates

A **ratio** is a numerical statement of how one variable relates to another. That is, it is a comparison of two numbers. Ratios are written as two numbers separated by a colon. For example, on older TV screens the ratio of width to height was 4:3. If the width is 40 cm, the height is 30 cm; if the width is 60 cm, the height is 45 cm; and so on. Modern, widescreen TVs have a ratio of 16:9.

A **rate** is a special kind of ratio that shows how long it takes to do something. For example, a very good athlete can run 10 000 m (10 km) in around 30 minutes. This is a rate of 1 km per 3 minutes, or 20 km per hour. Rate is much more meaningful than a simple count of how often something occurs. If you were investigating the effect of exercise on breathing, counting a person's breaths would be meaningless unless you knew how many breaths there were in a given time. That is, you need to know the rate in breaths per minute.

Percentages

'Per cent' means 'per hundred'. Percentages are used to express how large one variable is in relation to another. For example, if a breakfast cereal is labelled as containing 1.5% fat, it means that 100 g of the cereal contains 1.5 g of fat.

In Western Australia in 2011, males aged 15 to 19 years made up 6.7% of the population; females of the same age made up 6.4%. This means that for every 100 people in the population, 6.7 (or 67 per thousand) are 15 to 19 year-old males and 6.4 (64 per thousand) are 15 to 19 year-old females.

Percentage change

Calculating a percentage increase or decrease is often a good way to understand changes in a variable over time. For example, if a person weighing 100 kg lost 10 kg after dieting for six months, we could say that the person had lost 10% of their body weight as a result of the diet. If another person weighing 120 kg lost 13 kg after six months on the same diet, the percentage decrease would be 10.8%. Percentage change is helpful in making such comparisons.

To calculate percentage change:

1. *subtract* the old value (120 kg) from the new value (107 kg)
2. *divide* by the old value (120 kg)
3. *multiply* the result by 100 and add a per cent sign (%) to it.

This can be written as a formula:

$$\text{Percentage change} = \frac{\text{New value} - \text{Old value}}{\text{Old value}} \times 100$$

If the percentage change is positive, it indicates an increase; if the change is negative, it indicates a decrease.

Frequency

Frequency is the number of times an event occurs. For example, some students conducted a survey to find out how many drinks containing caffeine were consumed by the members of their class in a two-day period. The table of data they collected is called a **frequency distribution** or **frequency table**. A frequency table summarises the data by showing how often the variable in question occurs (Table 1.1). Frequencies can also be presented graphically as a **histogram** (Figure 1.8).

TABLE 1.1 Frequency table showing number of caffeine drinks consumed by students in a Year 12 class over a two-day period

NUMBER OF DRINKS CONSUMED	NUMBER OF STUDENTS
0	3
1	0
2	3
3	7
4	6
5	3
6	1
7	2

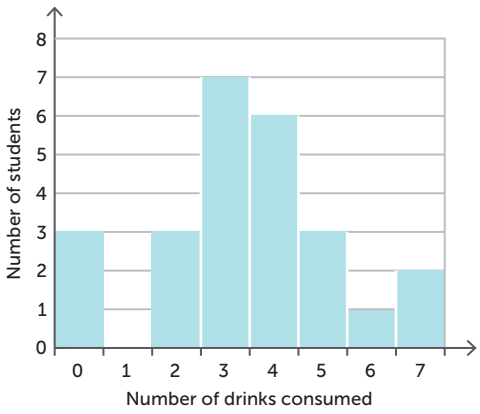

FIGURE 1.8 Histogram showing number of caffeine drinks consumed by students in a Year 12 class over a two-day period

Presentation of data

Tables

A convenient way to present numerical data is in the form of a table. The table of results for the students' survey of the number of caffeine drinks consumed in a Year 12 class over a two-day period could look something like Table 1.1.

Notice that the table follows these guidelines:

- It has a title. The title should state the variables represented by the data; in this case, the number of drinks consumed and the number of students are the two variables.
- The data is presented in columns. Usually the independent variable (in this case, the number of drinks consumed) is in the left column and the dependent variable (number of students) is in the right column. This rule is not always applied – it is more important that the table be easily understood.
- Each column has a heading and, where appropriate, the heading must state the units in which the data has been measured.

Graphs

Graphs are a pictorial way of presenting numerical data. A graph shows how changes in one variable affect another variable. From a graph it is easy to see any trends in the data. It is also possible to predict what the values would have been between the points plotted (**interpolation**), or the trend beyond the data shown in the graph (**extrapolation**).

Types of graphs

When drawing any graph, the following rules must be observed.

- The graph should have a title that states the two variables shown on the graph.
- The independent variable is plotted on the horizontal axis and the dependent variable is plotted on the vertical axis.
- Each axis is labelled with one of the variables and the units in which it is measured.
- Equal intervals of units are used on each axis.

The most commonly used graphs are line graphs (for continuous data), histograms (for frequencies) and, column graphs and bar graphs (for discrete data).

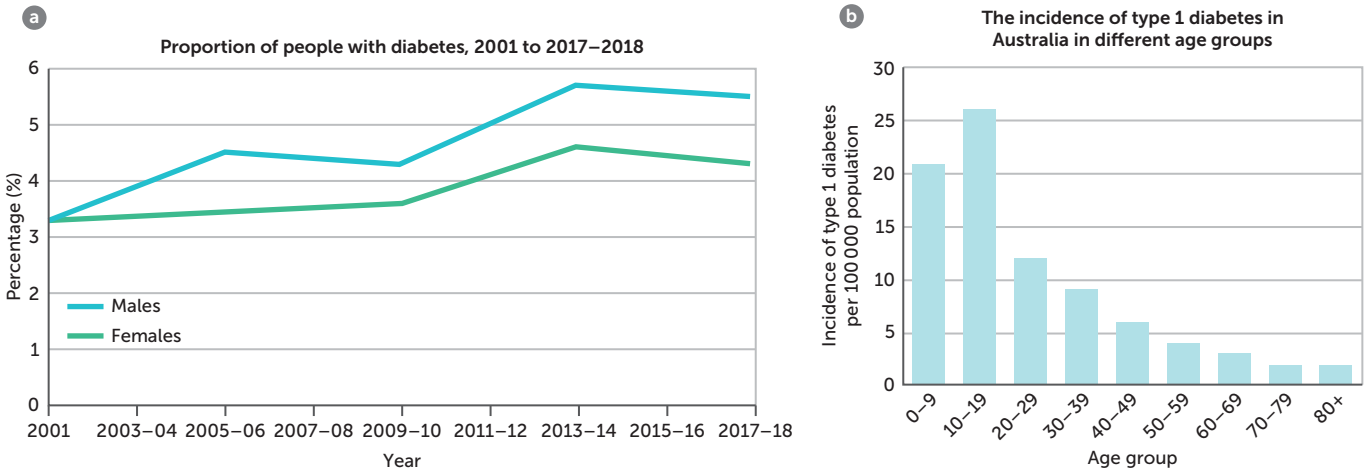

FIGURE 1.9 a Line graph; **b** Column graph. Data source a: Australian Bureau of Statistics, 4364.0.55.001 – National Health Survey: First Results, 2017–18. CC-BY 4.0 (https://creativecommons.org/licenses/by/4.0/); Data source b: Australian Institute of Health and Welfare (AIHW) CC-BY 3.0 (https://creativecommons.org/licenses/by/3.0/au/)

Key concept

Data is represented in tables and graphs and analysed to identify trends and patterns.

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Models

In science, a **model** is a simplified representation of an idea or process. Figure 1.5 (page 7) is a model of the scientific method, where it is being applied to the steps a doctor may take in trying to diagnose an illness. Once a model has been developed it can be applied to a number of situations. The model for the scientific method can be applied to most scientific investigations.

Figure 2.2, in Chapter 2 (page 30), is a model showing in simple diagram form how hormones may affect the functioning of a cell. The stimulus–response–feedback model shown in Figure 5.3, in Chapter 5 (page 106), is a model that can be applied to the regulation of body temperature, blood glucose and many other situations.

A model may be a diagram, a flow chart or a physical model such as a model of the atoms in a protein. Scientific models often have to be modified as new data is collected.

Flow charts

A flow chart is a diagram that shows the steps involved in a process. The steps are usually shown in boxes and the sequence of steps is indicated by arrows. Flow charts are very useful in summarising and visualising the steps in a complex process.

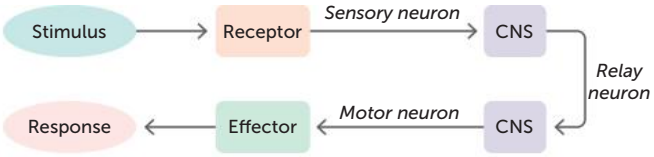

FIGURE 1.10 Flow chart for a reflex

Reference to the work of others

An extensive examination of the literature at the start of an investigation allows the researcher to fully grasp the available information relating to the problem under consideration. This review also allows the results to be seen in the context of what is already known. Research done by others can also be used to support or confirm what has been discovered in the investigation. Demonstrating how your findings relate to what is already known will give credibility to your research and will add to the body of knowledge on the subject under review.

WS

1.1 Scientific investigations

Questions 1.2

RECALL KNOWLEDGE

1 Arrange the steps of the scientific method in the correct order.
- Report on the investigation.
- Recognise a problem and define a question.
- Draw conclusions about whether the hypothesis was supported or disproved.
- Test the hypothesis using an experiment.
- Analyse and interpret the data collected from the experiment.
- Propose a hypothesis – a possible explanation for the problem.
- Collect as much information as possible relating to the problem.

2 List four procedures or devices that increase safety during an investigation. For each one, describe a risk that it would be used for.

3 Define 'ethics'.

4 Describe the principle of informed consent with regards to investigations involving humans.

5 Match the description with the relevant term.

REPETITION	During an experiment, the scientist completed five trials for each variation of the independent variable.
REPLICATION	Five different scientists followed the same method to test the same hypothesis.

APPLY KNOWLEDGE

6 Explain why an investigation may disprove a hypothesis but not prove it.

7 Literature review is an important component of a scientific investigation, as it allows scientists to assess methods used by others. Explain why this improves an investigation.

8 A group of students was testing the following hypothesis: 'Drinking caffeine increases focus while studying.' The students tested 20 Year 12 students, with five girls and five boys drinking a can of cola prior to studying and five girls and five boys drinking the same volume of water prior to studying. The time each student remained focused during their study was recorded.

- **a** State the independent variable for the investigation.
- **b** State the dependent variable for the investigation.
- **c** Describe the control group.
- **d** Rewrite the hypothesis to better reflect what was tested.
- **e** Was the investigation valid? Justify your answer.

9 During an investigation about the effect of different types of exercise, the following pulse rates, in beats per minute, were recorded prior to exercise.

54 65 62 58 60 66 84 57 61 65 59 63

- **a** Calculate the mean for the data.
- **b** Identify any outliers in the data.
- **c** State the median pulse rate.
- **d** State the range for the data.
- **e** During exercise, the mean pulse rate was 96 beats per minute. Calculate the percentage increase in pulse rate due to exercise.

10 The average heights of males of different ages are shown in the table.

AGE	AVERAGE HEIGHT (MALE) (CM)
20–24	164.44
25–29	164.32
30–34	163.59
35–39	163.59
40–44	163.31
45–49	163.50
50–54	162.93
55–59	162.16
60–64	161.21
65–69	160.46
> 70	158.30

Construct an appropriate graph to represent the data.

1.3 REPORTING ON SCIENTIFIC INVESTIGATIONS

When an investigation has been completed, the findings need to be made known to others. This is usually done by a written report. Reports are a very important part of communication in science. Scientists inform others of their research by publishing a report in a scientific journal. There are thousands of scientific journals, some of which deal with a very narrow field of science. Examples are *Nature, Science, Journal of Musculoskeletal Research* and *Journal of Genetics*.

The editors of scientific journals use a process called **peer review** to make sure that the report is worthy of publication. A submitted report is sent to one or more scientists who are experts in the field and who may or may not recommend publication. This process is important as it helps to keep scientific literature free of incorrect, bogus or misleading information.

A scientific report includes a description of an investigation, the results that were obtained and any conclusions that can be drawn from the results. The description of how the investigation was done must be sufficiently detailed to allow other scientists to repeat the experiment. It is common practice for scientists to repeat experiments that others have performed. If the results obtained are not the same as those for the original experiment, any conclusions that may have been drawn are questionable.

Scientific report format

Scientific reports generally follow a fairly standard format, as follows:

- title of the report and name of the author or authors
- introduction, stating the nature of the problem investigated and the hypothesis tested
- materials and equipment, listing the apparatus used, particularly any specialised items of equipment
- method, describing the exact method used to carry out the investigation
- results, often presented as tables, graphs, diagrams or photographs
- discussion, including comments about the results and the way they relate to the hypothesis that was tested as well as an evaluation of the investigation
- conclusion, summarising the most important parts of the discussion and stating whether or not the hypothesis was supported
- further research, as scientific investigations often raise more questions than they answer – many reports suggest areas that need further investigation
- references, which list any reports, books, journal articles, websites or other sources of information referred to in the report
- acknowledgements, of people who helped with the investigation or of organisations that provided funds for the research.

The discussion

The most important, and longest, part of a report is usually the discussion. The discussion is about the results and the method used to obtain the results. The discussion needs to be very thorough and to address all aspects of the research.

A checklist of questions that could be answered in the discussion section is as follows.

- Were there any defects in the design of the investigation or in the procedure?
- Were any results different from those expected?
- How do the results fit into the broader context of what is already known about the topic?
- Are there any practical applications for the results?
- Do the findings relate to any earlier work in the same area?
- Did the results support the hypothesis, or did they indicate that the hypothesis was incorrect?
- Were there any limitations in the research?
- Could the investigation have been improved in any way?
- Were there any variables that could not be controlled?
- Was there any bias in the results?
- Is there any information available from other reliable sources that would support the results?
- Is there a need for further research to clarify any of the results?

This is not an exhaustive list of questions. When writing a report, you will be able to think of other points that need to be discussed.

Report writing
This Monash University website gives detailed advice on report writing for scientific investigations.

Report writing FAQ
This University of New South Wales website gives advice on report writing and links to other useful sites.

Key concept

Scientific reports are used to communicate information about investigations. These reports undergo peer reviews to ensure the validity of the processes and results.

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Case study of a scientific investigation

French scientist Louis Pasteur (1822–95) conducted hundreds of investigations. His achievements include showing that micro-organisms cause disease, developing vaccines for rabies and some animal diseases, showing that micro-organisms are responsible for fermentation, and showing how the development of micro-organisms could be prevented by boiling and then cooling a liquid. This last process became known as pasteurisation.

Pasteur's investigations followed the scientific method. We can use aspects of his work as examples of many of the points discussed here. We will focus on Pasteur's demonstration that spontaneous generation does not occur. Spontaneous generation is the idea that living organisms can develop spontaneously from non-living matter.

Alamy Stock Photo/Science history Images

FIGURE 1.11 Louis Pasteur

Activity 1.1
Researching for Mightypharm

Italian physician Francesco Redi had shown in 1668 that maggots develop from eggs laid by flies. Until then it was believed that maggots formed naturally from rotting meat. Another Italian, Lazzaro Spallanzani, demonstrated 100 years later that micro-organisms come from the air and that boiling can kill them.

Despite the work of Redi and Spallanzani, the belief persisted that micro-organisms could spontaneously develop in decaying organic matter. The French Academy of Sciences arranged a contest for scientists to disprove the idea of spontaneous generation. Pasteur took up the challenge in 1859. This is a good example of how scientific knowledge builds over time. First Redi, then Spallanzani and later Pasteur and many others were involved in debunking the idea of spontaneous generation.

Pasteur's reports
This website has one of Pasteur's research reports on the growth of micro-organisms, published in 1860.

Pasteur had a problem to be solved and his hypothesis was 'that micro-organisms occur in sterile culture medium only when exposed to contaminated air from the outside'. To test the hypothesis he began a series of meticulous experiments. He opened flasks of sterile broth in the streets of Paris and found that after a time there was abundant growth of micro-organisms in the broth. He opened flasks high in the Alps and the broth nearly always remained sterile. Variables other than the location of exposure were kept the same: the flasks were the same size and shape, with the same volume of the same type of broth. All flasks were opened for the same period of time and kept at the same temperature, and so on. From his results Pasteur was able to conclude that the flasks exposed in Paris became infected because of the large numbers of micro-organisms in the air. The flasks exposed in the Alps remained free of micro-organisms because there are fewer micro-organisms in the air at high altitude.

Another experiment that Pasteur performed involved placing broth in flasks and heating them to kill micro-organisms. Some of the flasks then had their necks heated and drawn out into a long S-shaped curve. The necks of control flasks were heated but left straight, allowing the air to access the broth. All flasks were left in the same location with their necks open to the air. After several weeks the broth in the flasks with straight necks had gone cloudy due to the activity of micro-organisms. Broth in the curved-neck flasks remained clear; the micro-organisms and dust in the air settled in the bend of the S-shaped tube and did not reach the broth in the flasks. This experiment confirmed Pasteur's earlier conclusion that the air contains micro-organisms. Pasteur summarised his findings in a report titled 'On the Organised Bodies that Exist in the Air. Examination of the Doctrine of Spontaneous Generation'.

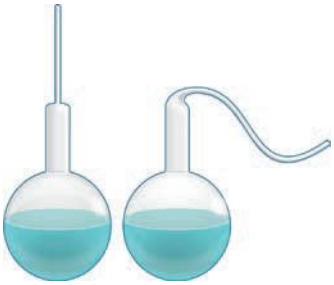

FIGURE 1.12
Types of flasks used by Pasteur in his experiment to demonstrate that spontaneous generation did not occur

Support for Pasteur's conclusions came from English physicist John Tyndall. He showed that sterile broth exposed to air but kept in a dust-free chamber remained sterile indefinitely. Tyndall and Pasteur were aware of each other's work – an example of one scientist producing evidence that supported the findings of another.


Alamy Stock Photo/INTERFOTO

FIGURE 1.13 John Tyndall

Despite the convincing evidence, the dispute over spontaneous generation continued. Many were not convinced, and Pasteur often had to defend his research. At a lecture in 1864 he said:

> *... there is now no circumstance known in which it can be affirmed that microscopic beings came into the world without germs, without parents similar to themselves. Those who affirm it have been duped by illusions, by ill-conducted experiments, spoilt by errors that they either did not perceive or did not know how to avoid.*

This situation persists in science today. The findings of scientists are subject to intense scrutiny by others and are often the subject of criticism – sometimes warranted, sometimes not. One reason for writing reports and presenting papers at conferences is so that other experts can examine the results.

Ideas about spontaneous generation were not finally laid to rest until 1876, when Pasteur and his assistant, Charles Chamberland, discovered that some bacteria produce spores that are resistant to high temperatures. These resistant spores accounted for the development of micro-organisms in cultures that had apparently been sterile for long periods. Some scientists had claimed that such development was the result of spontaneous generation. Spontaneous generation had finally been refuted 16 years after Pasteur's first convincing experiments and more than 200 years after Redi's research on the topic.

Activity 1.2
Validating Pasteur's experiment

Questions 1.3

RECALL KNOWLEDGE

1. State the reasons that peer reviews are an important part of scientific investigations.
2. Describe what you should include in each of the following sections of a scientific report:
   - **a** introduction
   - **b** method
   - **c** results
   - **d** conclusion
   - **e** acknowledgements.
3. Where did people believe micro-organisms came from prior to Pasteur's investigations?

APPLY KNOWLEDGE

4. The largest part of a scientific report is the discussion. The information presented in the discussion can be classified as:
   - discussing the results and their implications
   - evaluating the results
   - evaluating the method.

   Classify each of the following according to these options.
   - **a** Were there any defects in the design of the investigation or in the procedure?
   - **b** Were any results different from those expected?
   - **c** How do the results fit into the broader context of what is already known about the topic?
   - **d** Did the results support the hypothesis, or did they indicate that the hypothesis was incorrect?
   - **e** Could the investigation have been improved in any way?
   - **f** Were there any variables that could not be controlled?
   - **g** Was there any bias in the results?
5. Explain how Pasteur's investigation using broth in flasks with different necks was able to support the hypothesis 'that micro-organisms occur in sterile culture medium only when exposed to contaminated air from the outside'.
6. Discuss the importance of Tyndall and Chamberland's work supporting Pasteur's in disproving the idea of spontaneous generation.

CHAPTER 1 ACTIVITIES

ACTIVITY 1.1 Researching for Mightypharm

Researchers working for the pharmaceutical company Mightypharm were extracting chemicals from a new species of toadstool discovered in the rainforests of Brazil. Several of the chemicals were compounds that had never been found before. The researchers decided that the new compounds might have the potential to be used as antibiotics in the treatment of human bacterial infections.

Imagine that you are one of the Mightypharm researchers and your task is to test the new compounds with the goal of eventually producing an antibiotic that can be used to treat bacterial infections in human patients.

- *Stage 1:* Propose a hypothesis linking the two variables (chemical compounds and effect on bacteria). Describe how you would test the hypothesis to find out whether any of the compounds are effective in killing bacteria. Make your description detailed enough for someone else to follow and carry out the same tests that you propose. Describe how you would present your results and what sort of results would indicate that a compound had potential for use as an antibiotic.
- *Stage 2:* Suppose that one of the compounds tested in stage 1 showed promise as an antibiotic. Describe how you would test that compound on animals to find out whether it worked and whether there were any side effects from use of the compound. Make your description detailed enough for someone else to follow your procedure exactly, and remember that there are ethical considerations relating to the use of animals in research.
- *Stage 3:* The promising compound has successfully passed stages 1 and 2. Describe how you would carry out human trials on the compound. Also, describe how you would deal with any ethical issues that may arise.

In writing your descriptions of stages 1, 2 and 3 you may wish, or your teacher may ask you, to present your material as a paper to be published in a scientific journal. Refer to page 18 for the format of a scientific report.

Further investigation

You may wish to investigate how a prescription drug currently in use was discovered, developed and marketed.

ACTIVITY 1.2 Validating Pasteur's experiment

You can repeat Pasteur's experiment in which he used flasks with S-shaped necks, to see whether you get the same results.

You will need

For each pair or group:

- beef stock cubes
- filter funnel and filter paper
- four 100 mL conical flasks
- four one-hole stoppers
- straight glass tubing and S-shaped glass tubing
- source of heat (hot plate or Bunsen burner)

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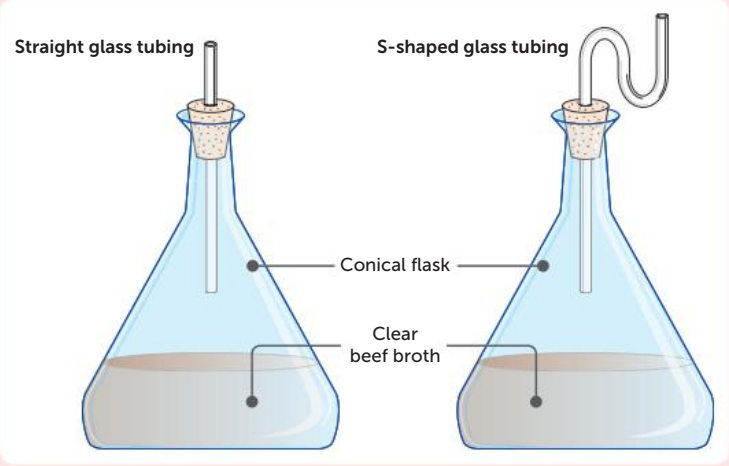
What to do

1. Make a broth using the beef stock cubes.
2. Filter the broth so that it is clear.
3. Place equal volumes of broth in each of the four flasks.
4. In two flasks, place a stopper with straight glass tubing and in the other two flasks place a stopper with S-shaped tubing.
5. Gently boil the broth in each flask for 15 minutes.
6. Leave the flasks in a warm place and check every couple of days for evidence of the growth of micro-organisms, such as cloudiness, a scum or mould on the surface of the liquid, or bubbles. You may need to leave your flasks for several weeks before any changes are apparent.
7. At the conclusion of the investigation do not open any of the flasks. They must first be autoclaved at 120°C for 20 minutes under 100 kPa pressure to destroy any micro-organisms.



Studying your results

1. Describe your results, giving a description of the broth in each flask.
2. Combine your results with those of other groups in the class. Explain the advantage of combining results.
3. Were your results similar to Pasteur's? Were the class results similar to Pasteur's?
4. If your results were not similar to Pasteur's, can you suggest any explanation?
5. If you were to repeat the experiment, how could you improve it?

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CHAPTER 1 SUMMARY

- Scientists use a range of different methods to acquire new knowledge, including observations, controlled experiments, surveys, trial and error, case studies and longitudinal studies.
- Controlled experiments involve changing one variable to identify the effect it has on the results.
- Surveys collect data from a large number of subjects to identify patterns.
- In science, trial and error involves testing a wide range of possibilities to find solutions.
- Case studies and longitudinal studies involve studying a particular person or situation. Longitudinal studies are conducted over a longer period than case studies.
- The scientific method provides a systematic approach to investigations.
- The scientific method involves identifying a problem, collecting information, proposing a hypothesis, testing the hypothesis, analysing the data and drawing a conclusion.
- Literature reviews survey material already written about the topic. This allows scientists to identify what is already known, assess research methods and relate findings to previous knowledge.
- It is always important to predict and address any safety issues for an investigation.
- Any investigations involving humans must adhere to ethical standards. This includes voluntary participation, informed consent, no risk of harm and confidentiality.
- Ethical principles must also be applied when working with animals.
- In a valid investigation, the independent variable is changed to determine its effect on the dependent variable. All other variables are controlled.
- Accurate data is close to the actual measurement. It is obtained by using the appropriate equipment, which has been calibrated correctly.
- Investigations are repeated and replicated in order to ensure the results remain the same.
- Reliable results occur when reliable measuring instruments are used and the same method is repeated.
- Data may be qualitative (descriptive) or quantitative (numerical), and can be represented in tables and graphs.
- An error, or a deviation from the correct value, is not necessarily a mistake in a scientific investigation. It can be due to a variation in the subject or measuring process.
- Data can be processed to determine the mean, outliers, median, range, ratio or rates, percentage change or frequencies.
- Models and flow charts can be used to represent ideas or processes.
- Peer reviews of scientific investigations ensure that they are valid.
- Scientific reports are used to communicate the process and findings from investigations.

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CHAPTER 1 GLOSSARY

Accuracy How close a measurement is to the true value

Arithmetic mean Often called the mean; the total measurements in a group divided by the total number of measurements

Average The total measurements in a group divided by the total number of measurements

Case study An in-depth investigation of one particular person or situation

Controlled experiment An experiment in which there are two almost identical set-ups; the only difference between them is the one variable being tested

Controlled variable A factor kept the same for both the control and the experimental groups in an experiment

Dependent variable In an experiment, the factor that changes in response to changes made to the independent variable; also called the responding variable

Ethical behaviour Behaviour that follows a set of moral principles or values

Ethics A set of moral principles or values

Extrapolation An estimation beyond the range of the original data

Frequency The number of times an event occurs

Frequency distribution *see* frequency table

Frequency table A summary of the data showing how often the variable in question occurs

Graph A pictorial way of presenting numerical data; shows how changes in one variable affect another variable

Histogram A graph to represent the frequency distribution of data

Hypothesis A statement of the relationship between the independent and dependent variables that is testable

Independent variable In an experiment, the factor being investigated; the factor deliberately changed to determine its effect; also called the experimental variable or the manipulated variable

Interpolation An estimation within the range of the original data

Literature review A survey of the material that has been written about a subject under consideration

Longitudinal study A study conducted over a long period of time; may be carried out over years or even decades

Mean *see* arithmetic mean

Measurement error The difference between a measurement and the true value of what is being measured

Median The mid-point of a set of numbers

Model A simplified representation of an idea or a process; may be a diagram, a flow chart, a simplified description of a complex situation or a physical model such as a model of a cell; examples are the stimulus–response–feedback model and the lock and key model for enzyme action

Observation The process of using the senses to acquire information

Outlier A measurement well beyond the range of other measurements in a set

Peer review The evaluation of work by people with similar skills and knowledge

Qualitative data Observations that do not involve numbers or measurement

Quantitative data Data expressed in numbers; usually involves measurement

Range The difference between the highest and lowest measurements in a group

Rate A ratio that shows how long it takes to do something

Ratio A numerical statement of how one variable relates to another; written as two numbers separated by a colon

Reliability The extent to which an experiment gives the same result each time it is performed

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Repetition Doing the same experiment many times

Replication Having a number of identical experiments running together, or performing the experiment on a large number of subjects at the same time

Scientific method A process of conducting valid investigations

Secondary data Data collected by someone other than the person using the data

Survey The systematic collection, analysis and interpretation of information about a particular question or series of questions; usually designed so that data is collected from a large number of subjects

Trial and error A problem-solving method in which one attempt to solve the problem is followed by another; each trial is recorded and the results allow the investigator to gradually home in on the solution

Uncontrolled variable A variable that could not be kept the same for the control and the experimental groups in an experiment

Validity The extent to which an experiment tests what it is supposed to test

Variable Any factor that may change during an experiment

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CHAPTER 1 REVIEW QUESTIONS

Recall

1 What is a controlled experiment?
2 List four principles that must be satisfied if an investigation is to be ethical.
3 What is a literature review and what are some of the reasons for carrying out such a review?
4 Describe how you would calculate the mean of a set of measurements.
5 What are outliers? Should outliers be excluded when drawing conclusions from a set of data?
6 Describe what a peer review is and why they are used.
7 Describe some of the points that should be included in the discussion section of a scientific report.
8 What is an 'error' when discussing a scientific investigation?

Explain

9 Explain the difference between:
   a observations and surveys
   b longitudinal studies and case studies.
10 a What is a hypothesis?
   b Can a hypothesis be proved? Explain.
11 a Explain the difference between the dependent and the independent variable in an experiment.
   b Explain the difference between controlled and uncontrolled variables.
12 Use an example to explain the difference between the validity and the reliability of an investigation.
13 Explain the difference between qualitative and quantitative data.

Apply

14 Re-read the account of Florey's experiment in which he injected mice with penicillin (page 4). List the variables that Florey controlled in his experiment.
15 What did Albert Einstein mean when he said: 'No amount of experimentation can ever prove me right; a single experiment can prove me wrong'?
16 Identify the type of investigation that would be the best for finding a solution to the following problems. Explain the reasons for your choice in each case.
   a Can people taste the difference between two different brands of milk chocolate?
   b What proportion of students in your school are left-handed?
   c What is the ratio of males to females in your Human Biology class?
   d How has a particular person's growth rate changed from birth to age 15?
17 In addition to physical activity that is part of their job or daily routine, many people deliberately exercise by going to a gym or by walking or jogging. Describe how you would conduct a survey to find out the average amount of time the teachers at your school spend on deliberate exercise.
18 The table below shows the systolic blood pressure of students in a Year 12 Human Biology class.

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SYSTOLIC BLOOD PRESSURE OF YEAR 12 STUDENTS (mmHg)					
109	123	141	115	131	126
144	138	106	115	49	109
125	132	128	114	116	120
195	143	132	116	13	

a Are there any obvious outliers in the data in the table? If so, which are the outliers and why should they be regarded as outliers?

b Calculate the mean systolic blood pressure for the class, excluding any outliers.

c What is the range of blood pressures in the class?

d What percentage of students had a blood pressure of 130 mmHg or higher?

e The average systolic blood pressure for adults is 120 mmHg. What proportion of students have blood pressures above this average?

19 Researchers investigating the benefits of exercise in preventing heart disease studied the health outcomes for women after participating in an exercise program. They calculated the risk of heart disease at 0.18 with a confidence interval of 0.04 to 0.80 at the 95% confidence level. Explain what the data means.

Extend

20 In 2003, a team of Australian anthropologists discovered skeletal remains on the Indonesian island of Flores. One skeleton was of a small human with a small brain, and dating showed it to be 18 000 years old. The team claimed it was a new species of human and named it *Homo floresiensis*. Experts are divided on whether the discovery is a new type of human or whether there is some other explanation for the small stature and small brain. This is a good example of scientific debate about the meaning of data. Use the Internet to find out some of the hypotheses put forward to explain why the skeleton is really our own species, *Homo sapiens*.

21 A research method sometimes used by scientists is meta-analysis. Find out what is meant by 'meta-analysis' and give an example of an investigation that used this method of research.

22 Some controlled experiments are said to be 'double-blind' experiments. Find out what is meant by the term 'double-blind experiment' and give examples of how such experiments might be used.

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2 HORMONES HELP CONTROL THE BODY

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

» select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions

SCIENCE UNDERSTANDING

Endocrine system

» hormones can be lipid soluble and able to cross cell membranes to bind with and activate intracellular receptors, or water soluble and able to bind with and activate receptors on cell membranes, and require secondary messengers to affect cell functioning

» the hypothalamus, pituitary, thyroid, parathyroid, pancreas, thymus, gonads, pineal and adrenal glands are endocrine glands found in the human body

» hormones secreted from the hypothalamus, pituitary, thyroid, parathyroid, pancreas and adrenal glands are involved in homeostasis by affecting specific target organs

» the secretions of the pituitary gland are controlled by the hypothalamus through transport of hormones, either via nerve cells or the vascular link between them

Source: School Curriculum and Standards Authority, Government of Western Australia

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The body is composed of trillions of cells that are organised into tissues, organs and systems. All these structures must work together in a coordinated way. This coordination is achieved through the activities of the nervous system and the endocrine system.

- The nervous system exerts control by the transmission of nerve impulses to and from the various tissues.
- The endocrine system influences the activity of cells by the release of chemical messengers known as hormones.

Much of the work of the endocrine system is concerned with keeping the environment inside the body fairly constant. Maintaining a stable internal environment is known as **homeostasis**. In this chapter, we will discuss how the endocrine system maintains homeostasis and controls cellular activities through chemical messengers.

2.1 ENDOCRINE SYSTEM

The endocrine system is made up of the endocrine glands, which secrete hormones.

Endocrine glands

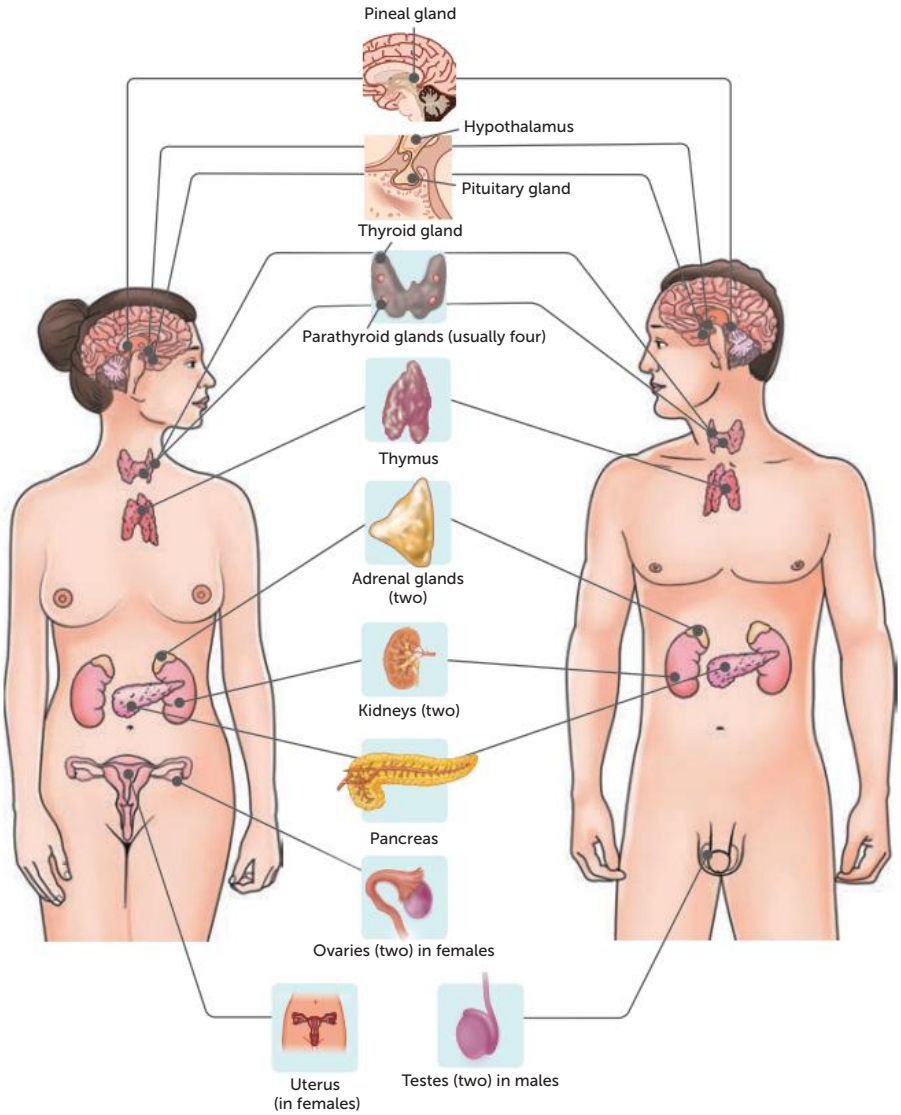

FIGURE 2.1
Endocrine glands

Endocrine system
This website contains more information about the endocrine system, the endocrine glands and hormones.

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There are two types of glands in the body.

- **Exocrine glands secrete** into a duct that carries the secretion to the body surface or to one of the body cavities. Sweat glands, mucous glands, salivary glands and the glands of the alimentary canal are examples of exocrine glands.
- **Endocrine glands** secrete hormones into the extracellular fluid that surrounds the cells that make up the gland. The secretion then usually passes into the capillaries to be transported by the blood. Endocrine glands are sometimes called ductless glands.

Hormones

Hormones are chemicals, secreted by endocrine glands, that are transported throughout the body in the blood. They change the functioning of cells by changing the type, activities or quantities of proteins produced. They are *not* enzymes; however, in many cases, hormones exert their influence by changing the activity of enzymes or their concentration. Hormones may:

- activate certain genes in the nucleus so that a particular enzyme or structural protein is produced
- change the shape or structure of an enzyme so that it is turned 'on' or 'off'
- change the rate of production of an enzyme or structural protein by changing the rate of transcription or translation during protein production.

Hormones are only able to influence cells that have the correct receptor for the hormone. Therefore, a hormone may affect:

- all the cells of the body
- only particular groups of cells, **target cells**
- only particular organs, **target organs**.

Hormones may be steroids, proteins or amines.

Steroid hormones

Steroid hormones, such as oestrogen, progesterone, cortisol and aldosterone, are lipid-soluble, meaning they do not dissolve in water. Once they are released into the blood, the hormones bind to transport proteins, enabling them to travel in the bloodstream. When they reach the target cells, the steroid hormones separate from the transport proteins and diffuse across the cell membrane. Inside the cell they work by combining with a receptor protein in the cytoplasm or nucleus. The hormone–receptor complex activates the genes controlling the formation of particular proteins. It does this by binding to the promoter section of a certain gene, stimulating (or inhibiting) transcription and, therefore, protein synthesis. Steroid hormones are slow to have an effect, but the effect is long lasting.

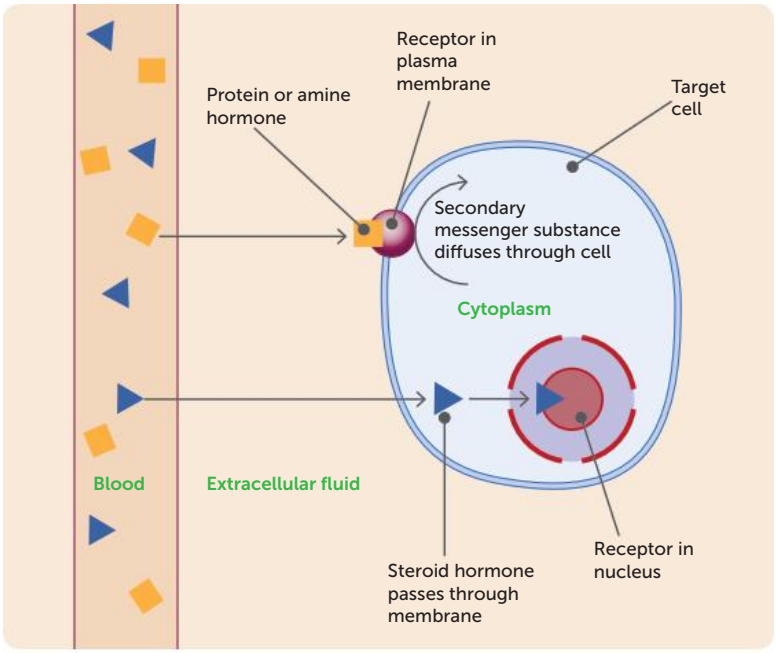


FIGURE 2.2 Hormones combine with receptors on the cell membrane or with receptors inside the cell

Protein and amine hormones

Protein and **amine hormones** are water-soluble. Because they are not lipid soluble, they are unable to diffuse across the cell membrane. Instead, they work by attaching to receptor proteins in the membrane of the target cell. The combination of the hormone with the receptor causes a secondary messenger substance to diffuse through the cell and activate particular enzymes. For example, the hormone insulin binds to a receptor protein and this leads to an increase in glucose absorption by the cell. Protein and amine hormones tend to be quick to cause a response; however, the effect is short lasting.

Hormone receptors

Receptor proteins are *specific*. Each type of receptor will bind with only one specific molecule. The lock and key analogy can be used to describe this interaction. The lock, the receptor protein, will only work with the correct key, the binding molecule.

There are a limited number of receptor proteins in the membrane of each cell. When each receptor is bound to a molecule, there can be no further increase in the rate of the cell's activity. For example, when each insulin receptor in the cell membrane is bound to insulin, the cell's rate of glucose uptake cannot increase any further, even if the amount of insulin increases. This means that *saturation* can occur; once all the receptor molecules are occupied by hormone molecules, the addition of more hormones does not produce any greater effect.

Different cells have different types and numbers of receptor proteins. This is why there is variation in the sensitivities of cells to hormones and other substances.

Steroid hormones
This website provides an animation of the way steroid hormones work.

Enzyme amplification

One hormone molecule does not cause the manufacture or activation of just one molecule of an enzyme – it activates thousands of molecules. This is achieved through a process called **enzyme amplification**. The hormone triggers a cascading effect in which the number of reacting molecules involved is increased hundreds or thousands of times for each step along the metabolic pathway. One hormone molecule could trigger the production of more than a billion enzyme molecules. Thus, a very small stimulus can produce a very large effect.

Hormone clearance

Once a hormone has produced the required effect, it must be turned off. This is done by breaking down the hormone molecules. Some hormones are broken down in the target cells, but most are broken down in the liver and the kidneys. The degraded hormones are then excreted in either the bile or the urine.

Control of hormone secretions

To maintain homeostasis, the amount of hormone produced by an endocrine gland must be very closely regulated. Any oversecretion or undersecretion of a hormone will cause the body to function abnormally.

Hormonal secretions are generally regulated by **negative feedback** systems whereby the response produced by the secretion of the hormone is the opposite of the stimulus that caused the secretion. Negative feedback systems will be covered in more detail in Chapter 5.

Key concept

The endocrine system is made up of the endocrine glands, which secrete steroid, protein or amine hormones that affect the functioning of the cell.

Questions 2.1

RECALL KNOWLEDGE

1. Where do exocrine glands secrete their products?
2. How do the products of endocrine glands move to their target cells?
3. List the three ways that hormones are able to change the functioning of cells.
4. Use a flow chart to show what happens to protein and amine hormones after they are secreted from the endocrine gland.

5 Describe the following properties of hormone receptors:
- **a** specific
- **b** saturation.

APPLY KNOWLEDGE

6 Explain why the receptors for steroid hormones are located inside the cell.

7 Predict what would happen if hormone clearance were unable to occur.

2.2 HYPOTHALAMUS AND PITUITARY GLAND

The hypothalamus and pituitary gland work together to control the functioning of many of the other glands. For this reason, the pituitary gland is often called the master gland. Combined, they can be thought of as the command centre for the body.

Hypothalamus

The **hypothalamus** has roles in both the nervous and endocrine systems and serves as a connection between the two systems. It regulates many of the basic functions of the body, such as body temperature, water balance and heart rate, in addition to increasing or decreasing the secretions of other glands.

The hypothalamus is located at the base of the brain, below the thalamus and above the pituitary gland. It is about the size of an almond.

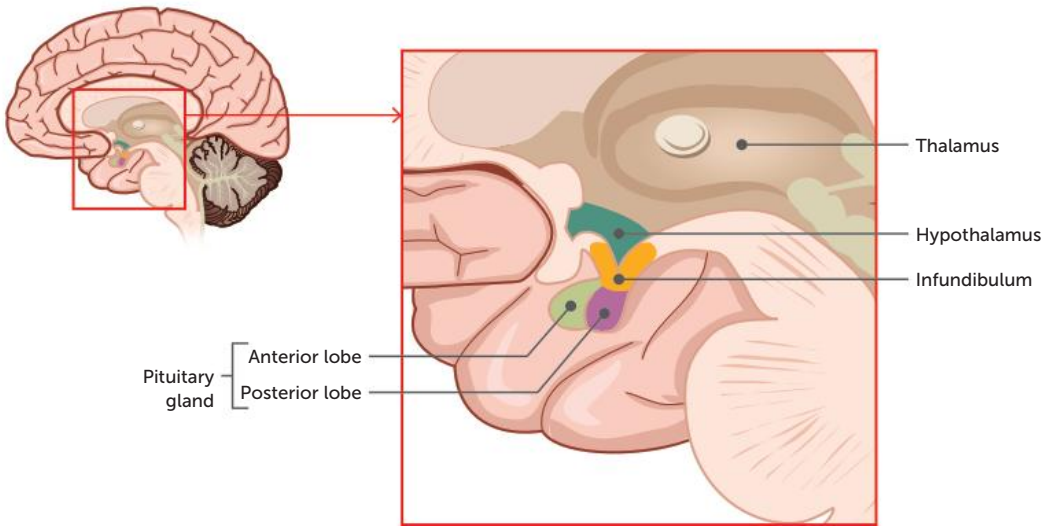


FIGURE 2.3 Location of the hypothalamus and the lobes of the pituitary gland

Hypothalamus and pituitary gland
This website provides more information on the relationship between the hypothalamus and the pituitary gland.

Pituitary gland and hypothalamus
This website gives more detailed information about the hypothalamus and pituitary glands.

Many of the functions of the hypothalamus are carried out through the pituitary gland.

- The hypothalamus secretes **releasing factors**, which stimulate the secretion of a hormone, or **inhibiting factors**, which slow down the secretion of a hormone. These factors travel through blood vessels to the anterior lobe of the pituitary gland, affecting the secretion of its hormones.
- Other hormones are produced by the hypothalamus and pass along the nerve fibres to the posterior lobe of the pituitary gland where they are then released.

Pituitary gland

The **pituitary gland**, or **hypophysis**, lies just under the hypothalamus and is joined to the hypothalamus by a stalk called the **infundibulum**. It is not much bigger than a large pea, about 13 mm in diameter, but it is absolutely vital to the normal functioning of the body.

The pituitary gland consists of an anterior lobe and a posterior lobe, each of which functions separately. The anterior (front) lobe has no nerves connecting it to the hypothalamus; rather, they are connected by a complex network of blood vessels lying in the infundibulum. The posterior (rear) lobe is joined to the hypothalamus by nerve fibres that come from nerve cell bodies in the hypothalamus and pass through the infundibulum to the posterior lobe. It is not a true endocrine gland because it does not secrete substances. Instead, it simply stores and releases hormones.

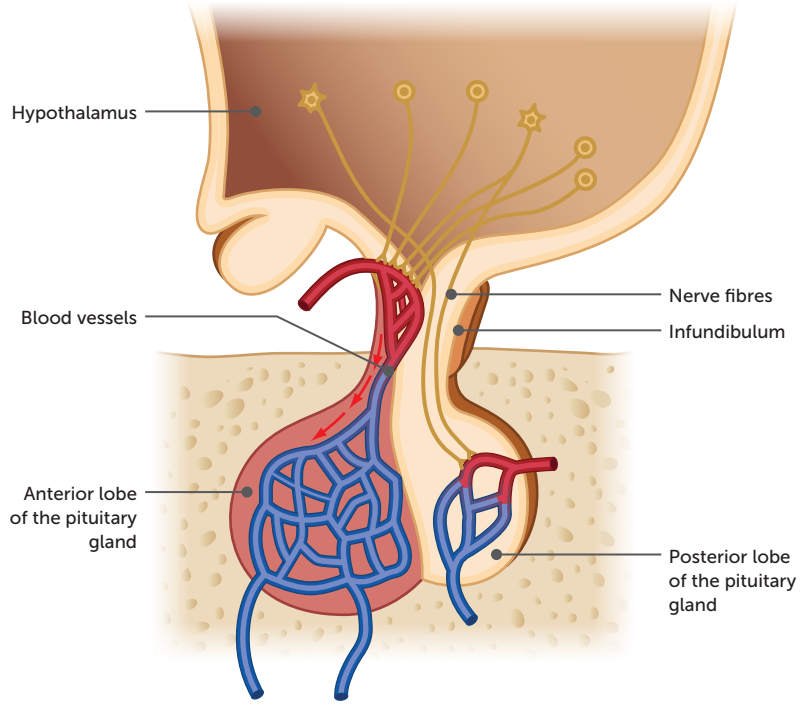


FIGURE 2.4 Blood vessels and nerves run through the infundibulum, connecting the hypothalamus to the lobes of the pituitary gland

Anterior lobe of the pituitary gland

The anterior lobe of the pituitary gland (the adenohypophysis) releases a number of hormones that regulate a great range of bodily activities. Secretions of the anterior lobe are in turn controlled by releasing and inhibiting factors secreted by the hypothalamus.

The following hormones are released by the anterior lobe of the pituitary gland:

- **Gonadotropins** are hormones that affect the gonads, the ovaries and testes. **Follicle-stimulating hormone (FSH)** stimulates development of the follicles that contain eggs in the ovary of the female. In the male, FSH stimulates the production and maturation of sperm in the testes. **Luteinising hormone (LH)** works with FSH in the female to bring about ovulation and to form a structure called the corpus luteum after ovulation. In the male, LH stimulates interstitial cells in the testes to secrete male sex hormones.
- **Growth hormone (GH)**, or somatotropin, stimulates body growth, particularly growth of the skeleton. It increases the rate at which amino acids are taken up by cells and built into proteins. GH is secreted throughout life as it helps to maintain the size of organs once maturity is reached.
- **Thyroid-stimulating hormone (TSH)**, or thyrotropin, stimulates production and release of hormones from the thyroid gland.
- **Adrenocorticotropic hormone (ACTH)**, or adrenocorticotropin, controls production and release of some of the hormones from the cortex of the adrenal glands.
- **Prolactin (PRL)**, or lactogenic hormone, works with other hormones to initiate and maintain milk production in females.

Posterior lobe of the pituitary gland

The posterior lobe of the pituitary gland (the neurohypophysis) releases the hormones oxytocin and antidiuretic hormone, but neither is manufactured in the posterior lobe. Both hormones are produced in special nerve cells in the hypothalamus of the brain. These cells have long extensions that pass through the infundibulum to the posterior lobe. Hormones manufactured in the cells move down the extensions and are stored ready for release into the bloodstream. The release of the hormones is triggered by nerve impulses initiated in the hypothalamus and conducted along the cell extensions.

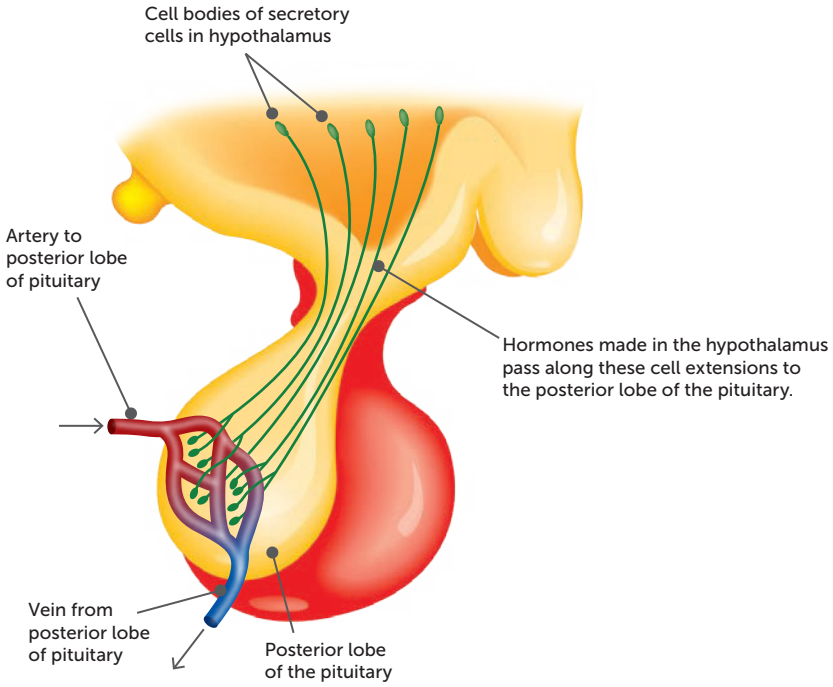


FIGURE 2.5 Hormones made in the hypothalamus are transported to the posterior lobe of the pituitary gland and can then be released into the bloodstream

Oxytocin (OT) stimulates contraction of the muscles of the uterus. It is released in large quantities during labour. Oxytocin also stimulates contraction of cells in the mammary glands, resulting in release of milk during breastfeeding.

Antidiuretic hormone (ADH), or **vasopressin**, causes the kidneys to remove water from urine that is forming. This water is returned to the bloodstream. In this way, ADH helps to retain fluid within the body. At higher concentrations, ADH can also cause constriction of small arteries, the arterioles. This is why its alternative name is vasopressin.

TABLE 2.1 Hormones released by the pituitary gland

HORMONE	TARGET ORGAN	MAIN EFFECTS
Anterior lobe of the pituitary gland		
Follicle-stimulating hormone (FSH)	Ovaries (females) Testes (males)	Growth of follicles Production of sperm
Luteinising hormone (LH)	Ovaries (females) Testes (males)	Ovulation and maintenance of corpus luteum Secretion of testosterone
Growth hormone (GH)	All cells	Growth and protein synthesis
Thyroid-stimulating hormone (TSH)	Thyroid gland	Secretion of hormones from the thyroid
Adrenocorticotropic hormone (ACTH)	Adrenal cortex	Secretion of hormones from the adrenal cortex
Prolactin (PRL)	Mammary glands	Milk production

HORMONE	TARGET ORGAN	MAIN EFFECTS
Posterior lobe of the pituitary gland		
Antidiuretic hormone (ADH)	Kidneys	Reabsorption of water
Oxytocin (OT)	Uterus Mammary glands	Contractions of uterus during childbirth Release of milk

Key concept

The hypothalamus is connected to the pituitary gland through nerves and blood vessels in the infundibulum. The hypothalamus communicates to the pituitary gland through these structures, influencing the release of hormones.

Questions 2.2

RECALL KNOWLEDGE

1. Describe the location of the hypothalamus.
2. One of the types of hormones that the hypothalamus secretes is releasing factors.
   - a. How do releasing factors reach their target cells?
   - b. What are the target cells of releasing factors?
   - c. What is the function of releasing factors?
3. What is the alternative name for the neurohypophysis?
4. Compare and contrast the anterior and posterior lobes of the pituitary gland.
5. List the gonadotrophins secreted by the anterior lobe of the pituitary gland and explain why they are classified as gonadotrophins.
6. List the hormones released from the posterior lobe of the pituitary gland.
7. Describe the target cells and function of each of the following hormones:
   - a. adrenocorticotropic hormone
   - b. prolactin
   - c. growth hormone
   - d. oxytocin
   - e. luteinising hormone
   - f. antidiuretic hormone
   - g. thyroid-stimulating hormone
   - h. follicle-stimulating hormone.

APPLY KNOWLEDGE

8. Explain why the pituitary gland is known as the master gland.
9. Predict what would happen if the infundibulum was severed.
10. Explain why the posterior lobe of the pituitary gland is technically not an endocrine gland.

2.3 OTHER ENDOCRINE GLANDS

Pineal gland

The **pineal gland** is found deep inside the brain. In children it is about the size of a pea. After puberty it gradually decreases in size. Its role remains something of a mystery, but it is known that it secretes the hormone **melatonin**, which is involved in the regulation of sleep patterns. Production of melatonin by the pineal gland is stimulated by darkness and inhibited by light.

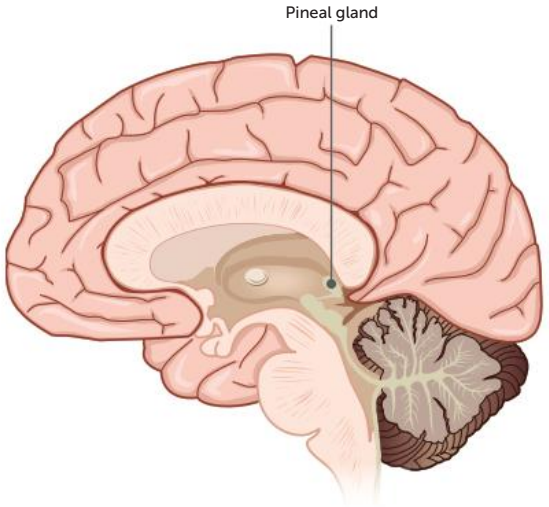


FIGURE 2.6 The pineal gland is located deep in the brain

Thyroid gland

The **thyroid gland** is located in the neck, just below the larynx. It consists of two lobes that lie on either side of the trachea and are joined by a narrow piece of tissue that lies across the front of the trachea.

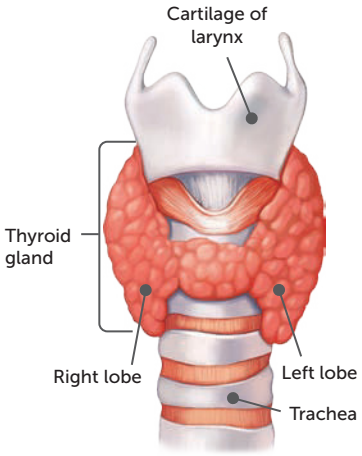
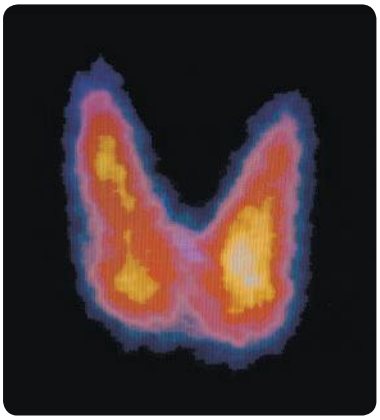


FIGURE 2.7 Location of the thyroid gland



Science Photo Library/Chris Priest

FIGURE 2.8 Scan of the thyroid gland, an endocrine gland in the neck. A radioactive tracer has been used to show the most active areas of the gland. These areas appear as yellow patches on the scan

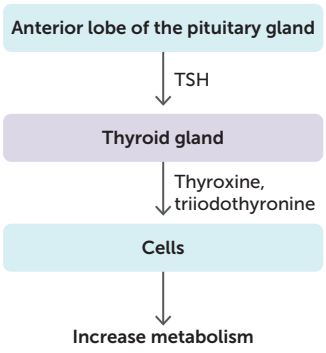


FIGURE 2.9 Thyroxine production

The follicular cells in the thyroid gland secrete two hormones in response to thyroid stimulating hormone: **thyroxine** (or T4) and **triiodothyronine** (or T3). The structure of both T3 and T4 is based on two molecules of the amino acid tyrosine. T3 has three iodine atoms attached while T4 has four iodine atoms attached. Thyroxine is much less active than triiodothyronine but lasts a lot longer. Approximately 80% of the thyroid hormones produced are thyroxine and only 20% are triiodothyronine. Once released, enzymes convert T4 into T3.

Thyroxine controls body metabolism by regulating reactions in which complex molecules are broken down to release energy, and other reactions in which complex molecules are synthesised from simple ones. The overall effect of thyroxine is to bring about the release of energy and, because some of the energy released is in the form of heat, to maintain body temperature.

The thyroid gland also plays a role in regulating the levels of calcium and phosphate in the blood through the release of **calcitonin** by C-cells. When the concentration of calcium in the blood increases, the thyroid gland releases calcitonin, which reduces the reabsorption of calcium by the kidneys and the breakdown of bone. If the concentration of phosphate in the blood becomes too high, calcitonin acts to move phosphate into bone and reduces its reabsorption by the kidneys. These actions allow calcium and phosphate concentrations to decrease.

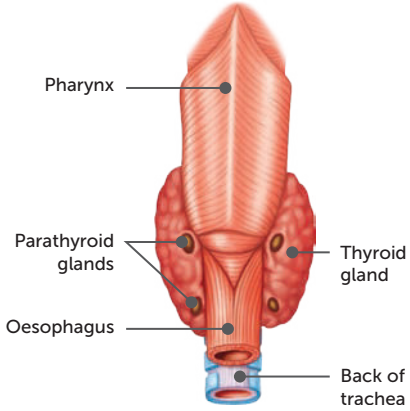


FIGURE 2.10 Location of the parathyroid glands (as seen from the rear of the body)

Parathyroid glands

The **parathyroid glands** are located in the rear surface of the lobes of the thyroid gland. There are usually four parathyroid glands, although some people have more. Each is about the size of a small pea.

The parathyroid glands secrete **parathyroid hormone** (**PTH**, or **parathormone**), which increases calcium levels in the blood and phosphate excretion in the urine.

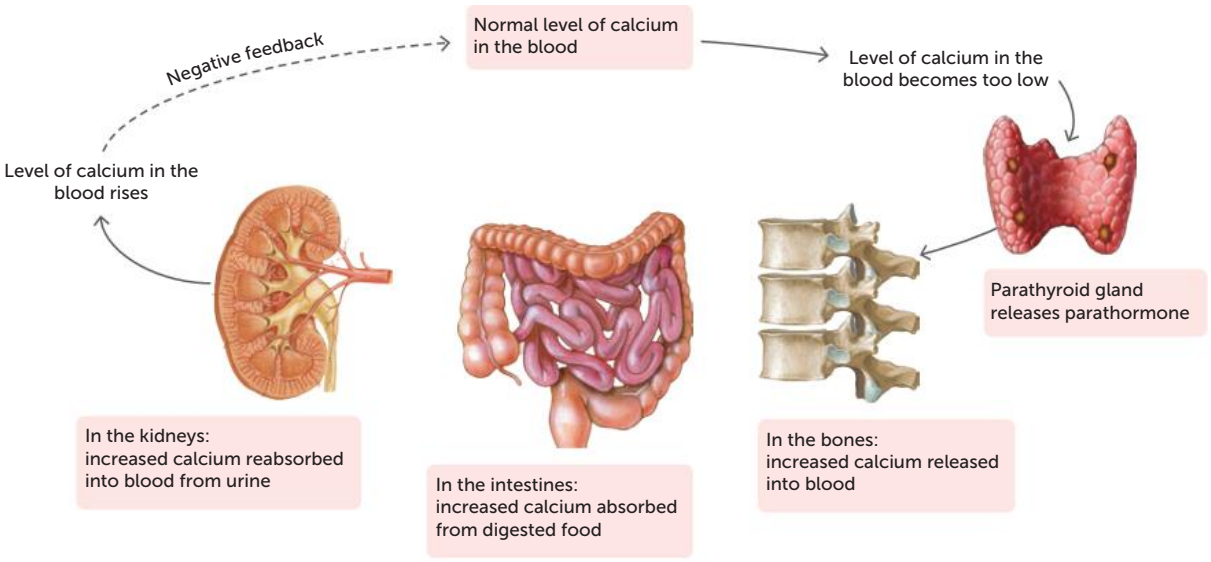


FIGURE 2.11 The regulation of blood calcium level by parathyroid hormone is a good example of negative feedback

Thymus

The **thymus** is located in the chest just above the heart and just behind the sternum. Like the pineal gland, the thymus is largest in infants and children, and begins to shrink after puberty. The thymus secretes a group of hormones called **thymosins**. These hormones influence the maturation of disease-fighting cells called T-lymphocytes. The role of T-lymphocytes will be discussed in Chapter 7.

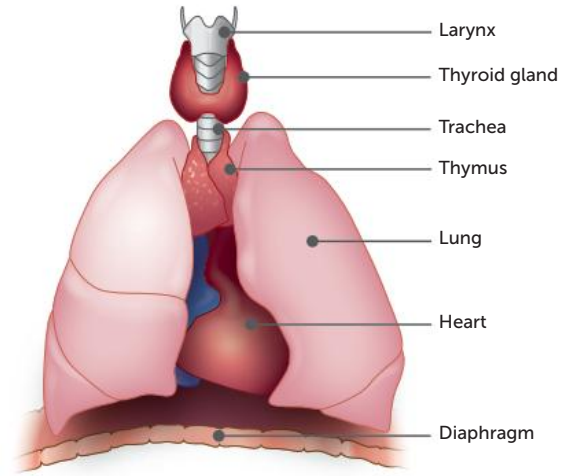


FIGURE 2.12 Location of the thymus

Adrenal glands

There are two **adrenal glands**, one immediately above each kidney. Each adrenal gland has an inner **adrenal medulla** and an outer **adrenal cortex**. These two parts are quite different in their structure and function. Thus, each adrenal gland is really two separate endocrine glands.

Adrenal medulla

The hormones produced by the adrenal medulla are adrenaline and noradrenaline.

- **Adrenaline**, also called **epinephrine**, has an effect similar to that of the sympathetic division of the autonomic nervous system. Adrenaline helps to prepare the body for reaction to a

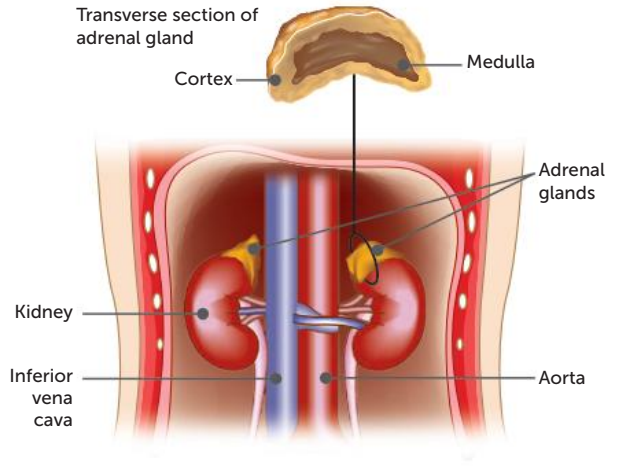


FIGURE 2.13 Location of the adrenal glands

threatening situation; that is, it is concerned with fight-or-flight responses. These responses will be discussed in more detail in Chapter 4.

- **Noradrenaline**, also called **norepinephrine**, has effects similar to those of adrenaline. In particular, it increases the rate and force of the heartbeat.

Adrenal cortex

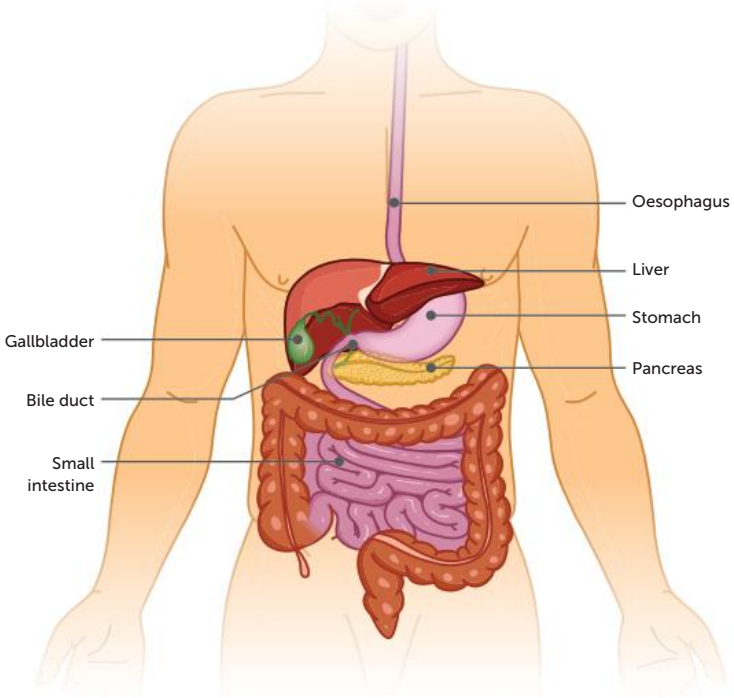
More than 20 different hormones are produced in the adrenal cortex and they are known collectively as **corticosteroids**. The two main ones are:

- **aldosterone**, which acts on the kidney to reduce the amount of sodium and increase the amount of potassium in the urine
- **cortisol**, which, with related hormones, promotes normal metabolism, helping the body to withstand stress and to repair damaged tissues.

Pancreas

The **pancreas** is a soft organ approximately 15 cm long. It lies just below the stomach and alongside the duodenum, the first part of the small intestine.

FIGURE 2.14
Location of the pancreas



The pancreas is both an exocrine gland and an endocrine gland. The exocrine part secretes digestive enzymes into the small intestine through the pancreatic duct. The endocrine part is made up of clusters of special cells called **islets of Langerhans** (also called **pancreatic islets**). These cells secrete two important hormones.

- **Insulin** is secreted by the beta cells of the islets of Langerhans. It reduces the amount of glucose in the blood (the blood sugar level). It does this by promoting the uptake of glucose from the blood by the cells of the body. In the liver, insulin causes the conversion of glucose to glycogen and fat; in skeletal muscles, it causes formation of glycogen from glucose; and in fat storage tissue, it causes glucose to be converted into fat. The level of secretion of insulin by the pancreas is determined by the amount of glucose in the blood and is controlled through a negative feedback system. This will be covered in more detail in Chapter 5.

Activity 2.1
Researching the discovery of insulin

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- **Glucagon**, secreted by the alpha cells of the islets of Langerhans, acts in the opposite way to insulin. It works to increase the blood glucose level, mainly by promoting the breakdown of glycogen to glucose in the liver. Glucagon also stimulates the breakdown of fat in the liver and in fat storage tissues.

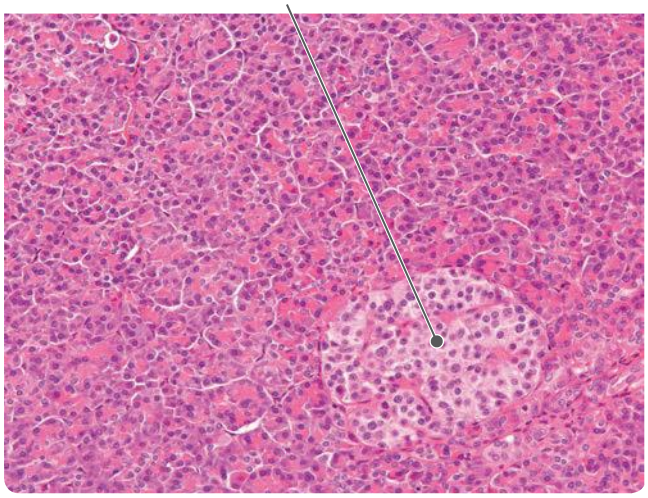

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FIGURE 2.15 Micrograph of islets of Langerhans within the pancreas

Gonads

The **gonads** are the testes and the ovaries. In Units 1 & 2, you learnt about their role in the reproductive system, with their production of sperm and eggs. They are also a part of the endocrine glands due to their production of hormones.

Androgens – for example, testosterone – are known as the male sex hormones. They are responsible for the development and maintenance of the male sex characteristics. In males, androgens are produced by the testes. Females also produce androgens in the ovaries, adrenal glands and fat cells. However, the levels produced in females are much lower than in males.

Oestrogens and **progesterone** are the female sex hormones produced by the ovaries. They stimulate the development and maintenance of the female sex characteristics. Together with the gonadotropic hormones of the pituitary, they also regulate the menstrual cycle and are involved in changes that occur during pregnancy.

Other endocrine tissues

In addition to the major endocrine glands discussed above, there are other tissues, many of which are not called endocrine glands, that secrete hormones. For example:

- The stomach and small intestine both secrete hormones that coordinate the exocrine glands of the digestive system.
- The kidneys secrete hormones, including **erythropoietin (EPO)** – a hormone that stimulates production of red blood cells by the bone marrow.
- The heart secretes a hormone that helps to reduce blood pressure.
- The **placenta** secretes a number of hormones during pregnancy that help to maintain the pregnancy, stimulate development of the foetus and stimulate the mother's mammary glands.

Major endocrine glands
This website provides further discussion of the major endocrine glands.

TABLE 2.2 Hormones produced by endocrine glands other than the pituitary gland

GLAND	HORMONE	TARGET CELLS	MAIN EFFECTS
Thyroid	Thyroxine and triiodothyronine Calcitonin	Most body cells Bones, kidney	Increases metabolic rate and therefore oxygen consumption and heat production Decreases calcium and phosphate levels in the blood
Parathyroid	Parathyroid hormone	Bones Kidneys	Increases level of calcium in blood
Thymus	Thymosins	T-lymphocytes	Stimulates development and maturation of T-lymphocytes

GLAND	HORMONE	TARGET CELLS	MAIN EFFECTS
Adrenal cortex	Corticosteroids, including:		
	Aldosterone	Kidneys	Increases reabsorption of sodium ions and excretion of potassium ions
	Cortisol	Most body cells	Promotes normal metabolism; helps the body deal with stress; promotes repair of damaged tissues
Adrenal medulla	Adrenaline and noradrenaline	Most body tissues	Prepares the body for fight-or-flight responses; reinforces the effects of the sympathetic nervous system
Pancreas	Insulin	Most body cells	Stimulates uptake of glucose; lowers blood glucose level
	Glucagon	Liver and fat storage tissues	Stimulates breakdown of glycogen and fat; increases blood glucose level
Testes	Androgens; e.g. testosterone	Many tissues	Stimulate sperm production, growth of skeleton and muscles, and development of male sexual characteristics
Ovaries	Oestrogens	Many tissues	Stimulate development of female sexual characteristics; regulate the menstrual cycle
	Progesterone	Uterus and mammary glands	Regulates menstrual cycle and pregnancy; prepares mammary glands for milk secretion

WS
2.1 Chemical messengers

Activity 2.2
Understanding endocrine dysfunction

Questions 2.3

RECALL KNOWLEDGE

1. Which endocrine gland secretes melatonin?
2. Describe the location of the thyroid gland.
3. Name the hormone secreted by the parathyroid gland and describe its function.
4. Name two endocrine glands that decrease in size with age.
5. Describe the location of the adrenal glands, and the arrangement of the adrenal medulla and adrenal cortex.
6. Match the hormone in the following table with the gland that produces it.

HORMONE	ENDOCRINE GLAND
adrenaline	adrenal cortex
thyroxine	pancreas
aldosterone	adrenal medulla
cortisol	ovaries
testosterone	testes
insulin	adrenal cortex
oestrogen	thyroid gland

APPLY KNOWLEDGE

7. Explain why people who suffer from hyperthyroidism (overactive thyroid) experience weight loss.
8. Explain how insulin and glucagon are able to keep blood glucose at the correct level.
9. Explain why oestrogen and progesterone are called the female hormones when they exist in both males and females.
10. Explain how the body is able to maintain a consistent level of calcium in the blood.

CHAPTER 2 ACTIVITIES

ACTIVITY 2.1 Researching the discovery of insulin

Insulin, a hormone secreted by the islets of Langerhans in the pancreas, was discovered in 1921–22 by researchers at the University of Toronto in Canada. The discovery is recognised as one of the greatest medical breakthroughs of all time.

Research into insulin continues to the present day. In the 1950s the full sequence of amino acids in the insulin molecule was determined, and in 1969 scientists worked out its three-dimensional structure.

Your task

1. Use the Internet to research the story of insulin's discovery by:
   - Frederick Banting
   - Charles Best
   - JJR Macleod
   - James Collip.

   For each scientist, include the method they used to isolate insulin.
2. In 2006 a research team at the CSIRO in Australia made another important discovery in the quest for a full understanding of how insulin works. Find out what the discovery was and its implications for the understanding of insulin.

ACTIVITY 2.2 Understanding endocrine dysfunction

Many disorders can be caused by an oversecretion or undersecretion from one or more of the endocrine glands. Some of these disorders are listed below.

- Acromegaly
- Addison's disease
- Androgen insensitivity syndrome
- Cushing's syndrome
- Exophthalmia
- Gigantism
- Goitre
- Graves' disease
- Myxoedema
- Phaeochromocytoma

Select one of these hormonal problems and use references to learn about:

- the endocrine gland and hormone involved
- the symptoms of the disorder
- whether the disorder is caused by an oversecretion or undersecretion of the hormone
- the treatment that is available for the disorder.

Write a short essay to show the cause-and-effect relationship between the information that you researched.

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CHAPTER 2 SUMMARY

- The nervous and endocrine systems are responsible for maintaining homeostasis.
- Endocrine glands secrete hormones into the extracellular fluid, where they diffuse into capillaries to be transported in the blood vessels.
- Hormones change the functioning of cells by altering the type, activity or quantity of proteins produced.
- Hormones affect target cells, which have specific receptors for the hormone.
- Lipid-soluble hormones (steroid hormones) diffuse through the cell membrane to bind to receptors inside the cell.
- Water-soluble hormones (protein and amine hormones) bind to receptors on the cell membrane. This triggers a secondary messenger in the cell.
- Hormone receptors are specific and can become saturated.
- One hormone can lead to the production of many enzyme molecules. This is known as enzyme amplification.
- Negative feedback systems control the secretion of hormones.
- The hypothalamus and pituitary gland work together, and control many other endocrine glands.
- Secretions from the anterior lobe of the pituitary gland are controlled by releasing and inhibiting factors from the hypothalamus. These secretions travel to the anterior lobe through blood vessels in the infundibulum.
- The anterior lobe of the pituitary gland secretes follicle-stimulating hormone, luteinising hormone, growth hormone, thyroid-stimulating hormone, adrenocorticotropic hormone and prolactin.
- The posterior lobe of the pituitary gland stores and releases hormones that are produced by the hypothalamus. They travel to the posterior lobe along nerve fibres in the infundibulum and are released due to nerve impulses from the hypothalamus.
- The posterior lobe of the pituitary gland releases oxytocin and antidiuretic hormone.
- The pineal gland secretes melatonin, which regulates sleep patterns.
- The thyroid gland releases thyroxine and triiodothyronine, which increase metabolism to release energy, including heat. It also secretes calcitonin, which decreases the level of calcium and phosphate in the blood.
- The parathyroid glands release parathyroid hormone, which increases the calcium level in the blood and phosphate excretion in the urine.
- The thymus secretes thymosins, which play a role in the maturation of T-lymphocytes.
- The adrenal medulla is the inner part of the adrenal gland. It secretes adrenaline and noradrenaline, which play a role in the fight-or-flight response.
- The adrenal cortex is the outer part of the adrenal gland. It secretes aldosterone, which regulates sodium and potassium excretion, and cortisol, which protects the body from stress.
- The endocrine cells in the pancreas form the islets of Langerhans. They secrete insulin, which decreases blood glucose levels, and glucagon, which increases blood glucose levels.
- Androgens, such as testosterone, are produced by the testes and are responsible for the male sex characteristics.
- Oestrogens and progesterone are produced by the ovaries. They are responsible for the female sex characteristics and help to regulate the menstrual and ovarian cycles.
- The stomach, kidneys, heart and placenta are also considered endocrine glands as they produce hormones.

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CHAPTER 2 GLOSSARY

Adrenal cortex The outer portion of an adrenal gland; secretes the hormones aldosterone and cortisol

Adrenal glands Two endocrine glands, one immediately above each kidney; each consists of an inner adrenal medulla and an outer adrenal cortex

Adrenal medulla The inner portion of an adrenal gland; secretes the hormones adrenaline and noradrenaline

Adrenaline A hormone secreted from the adrenal medulla that prepares the body for the fight-or-flight responses; also called epinephrine

Adrenocorticotropic hormone (ACTH) A hormone that controls the production and release of some of the hormones from the cortex of the adrenal glands

Aldosterone A hormone that acts on the kidney to reduce the amount of sodium in the urine and increase the amount of potassium

Amine hormone A hormone composed of an amino acid with modified groups

Androgen Any of the male sex hormones produced by the testes; responsible for the development and maintenance of the male sex characteristics

Antidiuretic hormone (ADH) A hormone produced by the hypothalamus and released by the posterior lobe of the pituitary gland that stimulates the kidneys to remove water from urine, thus reducing urine production; also known as vasopressin

Calcitonin A hormone secreted by the C-cells of the thyroid gland that decreases the concentrations of calcium and phosphate

Corticosteroid Any of a group of more than 20 hormones secreted by the adrenal cortex; the two main hormones are aldosterone and cortisol

Cortisol A hormone that, along with related hormones, promotes normal metabolism

Endocrine gland A gland that secretes hormones directly into adjacent tissue; also called a ductless gland

Enzyme amplification A series of chemical reactions in which the product of one step is an enzyme that produces an even greater number of product molecules at the next step

Epinephrine *see* adrenaline

Erythropoietin (EPO) A hormone that stimulates production of red blood cells by the bone marrow

Exocrine gland A gland that secretes into a duct that carries the secretion to the surface of the body cavities

Follicle-stimulating hormone (FSH) A hormone that stimulates the development of a follicle in the ovary

Glucagon A hormone secreted by the pancreas that increases blood sugar level

Gonadotropin A hormone that affects the sex organs

Gonads The testes and ovaries

Growth hormone (GH) A hormone that stimulates body cells to grow and multiply, especially the skeleton and skeletal muscles

Homeostasis The maintenance of a relatively constant internal environment despite fluctuations in the external environment

Hormone A chemical that is secreted by an endocrine gland and that affects the functioning of a cell or organ; often carried in the blood

Hypophysis An alternative name for the pituitary gland

Hypothalamus The part of the brain lying just below the thalamus and above the pituitary gland; controls many homeostatic mechanisms, such as body temperature, water balance and heart rate

Infundibulum The stalk-like structure that joins the pituitary gland to the hypothalamus

Inhibiting factor A hormone that slows the release of another hormone

Insulin A hormone, secreted by the pancreas, that decreases blood sugar level

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Islets of Langerhans Clusters of endocrine cells in the pancreas; secrete the hormones insulin and glucagon

Luteinising hormone (LH) A hormone that promotes final maturation of the ovarian follicle and the formation of the corpus luteum

Melatonin A hormone secreted by the pineal gland that regulates sleep patterns

Negative feedback Feedback that reduces the effect of, or eliminates, the original stimulus

Noradrenaline A hormone secreted from the adrenal medulla that has effects similar to that of adrenaline; in particular, it increases the rate and force of the heartbeat; also called norepinephrine

Norepinephrine *see* noradrenaline

Oestrogen A female sex hormone; develops or maintains female reproductive structures and regulates the menstrual cycle and pregnancy

Oxytocin The hormone that stimulates contraction of the muscles of the uterus

Pancreas A gland that lies just below the stomach; both an endocrine and exocrine gland; secretes digestive enzymes from the exocrine cells, and the hormones insulin and glucagon from endocrine cells

Pancreatic islet *see* islets of Langerhans

Parathormone *see* parathyroid hormone

Parathyroid gland One of four (usually) small glands about the size of a small pea embedded in the rear surface of the thyroid gland; secretes parathyroid hormone (PTH)

Parathyroid hormone (PTH) A hormone that controls calcium and phosphate levels in the blood; also known as parathormone

Pineal gland A small gland, about the size of a pea in children, found deep inside the brain; in adults it is just a tiny lump of fibrous tissue; the functions of the hormones it secretes have still not been identified

Pituitary gland An endocrine gland located below the brain; joined to the hypothalamus by a stalk called the infundibulum

Placenta The organ that supplies nutrients to, and removes wastes from, the foetus; also produces a number of hormones, including oestrogens and progesterone

Progesterone A female sex hormone produced by the ovaries; helps prepare the uterine lining for a fertilised egg; also prepares the mammary glands for milk secretion

Prolactin (PRL) A hormone that promotes milk production during and after pregnancy

Protein hormone A hormone consisting of a long chain of amino acids (a protein)

Releasing factor A hormone whose purpose is to control the release of another hormone

Secretion A useful substance produced and released by a gland or cell (noun); the process of producing and releasing a useful substance (verb)

Steroid hormone A hormone derived from the lipid cholesterol

Target cell A cell whose activity is affected by a particular hormone

Target organ An organ whose activity is affected by a particular hormone

Thymosin Any of a group of hormones secreted by the thymus that stimulate the immune system by helping the maturation of T-lymphocytes

Thymus An endocrine gland located in the chest just above the heart and behind the sternum; secretes a group of hormones called thymosins

Thyroid gland An endocrine gland, consisting of two lobes, located in the neck just below the larynx; secretes the hormone thyroxine

Thyroid-stimulating hormone (TSH) A pituitary hormone that stimulates production and release of hormones from the thyroid gland; also known as thyrotropin

Thyroxine A hormone secreted by the thyroid gland that regulates metabolism, growth and development

Triiodothyronine The more active thyroid hormone that increases the rate of metabolism, also known as T3

Vasopressin *see* antidiuretic hormone

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CHAPTER 2 REVIEW QUESTIONS

Recall

1 a Describe the endocrine system.
b Describe the relationship between endocrine glands and hormones.

2 a Define 'hormone'.
b List the different types of hormones.

3 Describe enzyme amplification and state why it is important.

4 The pituitary gland is sometimes described as the 'master gland' because it secretes hormones that regulate the activity of other endocrine glands. Describe the pituitary hormones that are involved in the control of other endocrine glands.

5 a What is a target organ?
b How do hormones get from their source to the target organ?
c Describe target organs/cells and the role of the following hormones.
i Oxytocin
ii Antidiuretic hormone
iii Adrenaline
iv Parathyroid hormone
v Insulin
vi Glucagon
vii Thyroxine

6 a Which gland produces thymosins, and what is the function of these hormones?
b Which gland secretes melatonin? What is the role of melatonin?

Explain

7 Explain the difference between endocrine and exocrine glands and give five examples of each.

8 Hormones are specific. Explain what this means and how it is achieved.

9 The hypothalamus and the pituitary gland are closely related. Describe their relationship in terms of:
a their location in the body
b the ways in which they function.

10 Hormones act by changing the functioning of a cell. Explain how they are able to do this.

11 Hormones that are lipid-soluble work in a different way from those that are water-soluble. Explain the difference and why it occurs.

12 Hormones secreted by the posterior lobe of the pituitary are not actually made in the posterior lobe. Explain the process of producing and releasing these hormones.

Apply

13 Explain why endocrine glands are sometimes called ductless glands.

14 Hormones affect the activity of their target cells. Explain why the addition of more and more hormone does not continue to increase the intensity or rate of the response.

15 Athletes have sometimes taken (illegally) the hormone erythropoietin in an effort to improve their performance. In what ways would this hormone improve sporting performance?

16 Construct a flow diagram similar to Figure 2.11 for the hormone ADH and its role in water balance. Include the role of feedback in your diagram.

17 Thyroid-stimulating hormone (TSH) is secreted by the anterior lobe of the pituitary gland. If a cancer patient had their thyroid gland removed, would you expect the level of TSH in the person's blood to rise or fall? Explain your answer.

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Extend

18 Many famous people have suffered from endocrine disorders.

- **a** John F Kennedy, President of the United States from 1960 until his assassination in 1963, suffered from Addison's disease. Consult references to see if you can find out some of Kennedy's medical history. How was he able to carry out his duties as President of the United States while having such a serious illness?
- **b** Napoleon Bonaparte is believed to have suffered from a disease of the hypothalamus that caused the pituitary gland to function abnormally. Because the anterior lobe regulates the functioning of the gonads and the adrenal and thyroid glands, these organs were also affected. See if you can find out the symptoms of Napoleon's disorder.
- **c** Akhenaton, an Egyptian pharaoh who lived 3500 years ago, is portrayed in statues made later in his life with feminine features – prominent breasts, hips wider than the shoulders, and a large amount of fat on the buttocks and thighs. It has been suggested that Akhenaton may have had a disorder of one of the endocrine glands. Which gland, or glands, could it have been, and what hormones could have been involved?

19 In an average person the thymus weighs about 35 g just before puberty, but by age 50 it has shrunk to around 12 g and by 75 to about 6 g. It has been suggested that this decline in size may be responsible for elderly people becoming more susceptible to disease. Research the thymus to find out:

- **a** how the role of the thymus was discovered
- **b** the role of the thymus in providing defence against disease.

20 New hormones are still being identified. One well-known example is the hormone leptin, discovered in 1994 through the study of obese mice. Leptin is secreted by fat storage tissues (adipose tissues). Find out:

- **a** how leptin was discovered
- **b** the target cells for leptin
- **c** the effect of the hormone.

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3 NEURONS COMMUNICATE QUICKLY

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including the collection of data related to homeostasis and the use of models of disease transmission, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- interpret a range of scientific and media texts, and evaluate models, processes, claims and conclusions by considering the quality of available evidence, and use reasoning to construct scientific arguments
- select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

SCIENCE UNDERSTANDING

Central and peripheral nervous system

- transmission of nerve impulses is via electro-chemical changes that occur at the generation of the impulse, the propagation of the impulse along the nerve fibre, and the transfer of the impulse across the synapse
- different receptors detect changes in the internal and external environments, including thermoreceptors, osmoreceptors, chemoreceptors and receptors for touch and pain
- the reflex arc is composed of specially structured neurons, including sensory, interneuron and motor neurons, to transmit information from the receptor to the effector to respond rapidly to stimuli
- the nervous and endocrine systems work together to co-ordinate functions of all body systems, but differ in terms of:
  - speed of action
  - duration of action
  - nature and transmission of the message
  - specificity of message

Source: School Curriculum and Standards Authority, Government of Western Australia

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The nervous system is one of the body's two communications systems. Along with the endocrine system studied in Chapter 2, it coordinates all our voluntary and involuntary actions. The nervous system receives and processes information from sense organs and brings about responses to the information received.

3.1 NERVE CELLS

Nerve cells, or **neurons**, are the basic structural and functional units of the whole nervous system. They are highly specialised cells perfectly designed for rapid communication of messages in the body.

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FIGURE 3.1 A coloured scanning electron micrograph of two nerve cells from the brain

Structure of neurons

Neurons vary in size and shape, but they all consist of a cell body and two different types of extension from the cell – the dendrites and the axon.

Cell body

The **cell body** is the part of the neuron that contains the nucleus and is responsible for controlling the functioning of the cell. Around the nucleus is cytoplasm containing the organelles that are found in most cells: mitochondria, endoplasmic reticulum, ribosomes and Golgi apparatus.

Dendrites

Dendrites are usually fairly short extensions of the cytoplasm of the cell body. They are often highly branched and they carry messages, or nerve impulses, into the cell body.

Axon

The **axon** is often a single, long extension of the cytoplasm. It usually carries nerve impulses away from the cell body. Although usually longer than the dendrites, the length of axons varies enormously. Those in the brain may be only a few millimetres long, while the axons that run from the spinal cord to the foot may be a metre or so in length.

At its end, the axon divides into many small branches. Each of these branches terminates at the **axon terminal**.

Myelin sheath

Most axons are covered with a layer of fatty material called the **myelin sheath**. The term **nerve fibre** is used for any long extension of a nerve cell, but usually refers to an axon. Those that have a myelin sheath are called **myelinated fibres** and those that do not are said to be **unmyelinated**.

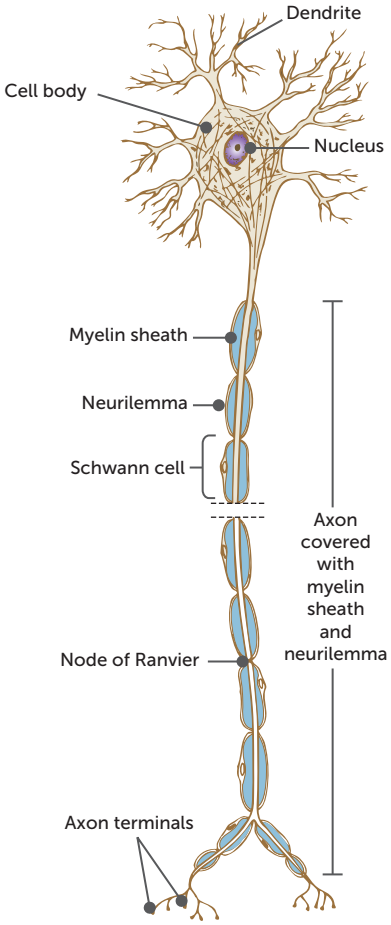

FIGURE 3.2 Structure of a typical neuron with a myelinated axon

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Outside the brain and spinal cord, the myelin sheath is formed by special cells called **Schwann cells**, which wrap around the axon. At intervals along the axon are gaps in the myelin sheath, called **nodes of Ranvier**.

The myelin sheath has three important functions:

- It acts as an insulator.
- It protects the axon from damage.
- It speeds up the movement of nerve impulses along the axon.

The outermost coil of the Schwann cell forms a structure called the **neurilemma** around the myelin sheath. This structure helps in the repair of injured fibres.

In the brain and spinal cord, the myelin sheath is produced by oligodendrocytes. The fatty nature of the myelin means that the areas containing myelinated fibres appear white and are called white matter. The areas made up of cell bodies and unmyelinated fibres are called grey matter due to their grey colour.

Activity 3.1
Creating a model of a neuron

Key concept

Nerve cells, called neurons, are composed of a cell body, axon and dendrites.

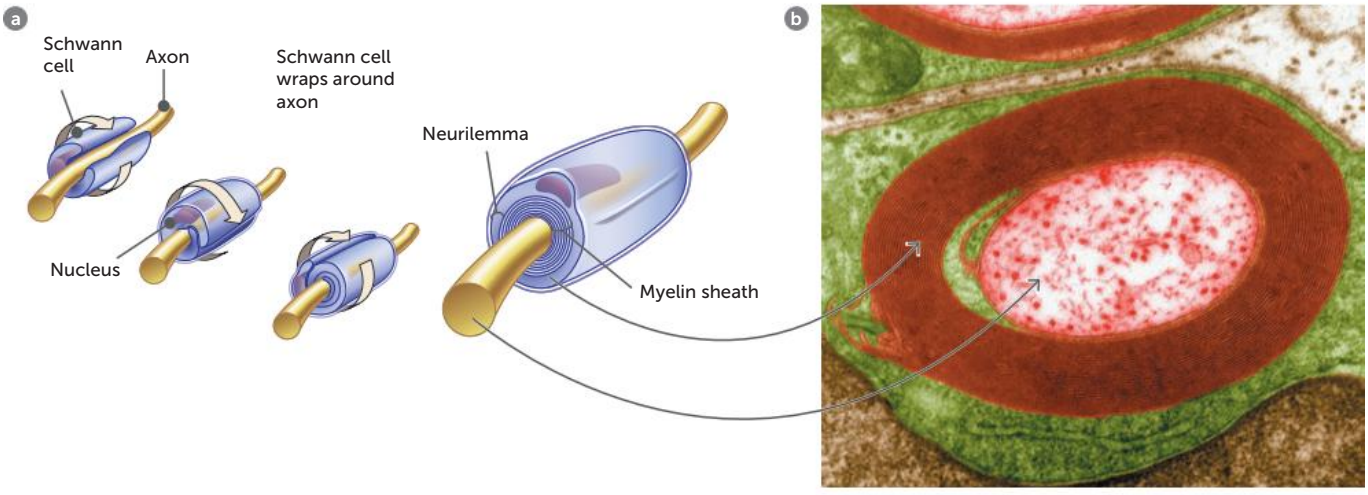

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FIGURE 3.3 a Schwann cells form the sheath of a myelinated axon by wrapping around the axon. **b** Coloured transmission electron micrograph of a transverse section of a myelinated axon. The myelin sheath (brown) is formed when Schwann cells wrap around the axon and deposit layers of myelin between each coil. The outermost layer (green) is the cytoplasm of the Schwann cell – the neurilemma

Synapses

Nerve impulses have to be passed from neuron to neuron. This usually occurs where the axon terminal of one neuron joins with a dendrite or the cell body of another. This junction is called a **synapse**.

The neurons do not actually physically touch at the synapse; instead, there is a small gap between them. Messages have to be carried across this gap, which occurs by the movement of chemicals called **neurotransmitters**.

A similar synapse exists where an axon meets a skeletal muscle cell. This tiny gap is called the **neuromuscular junction**.

Synapses and neuromuscular junctions will be discussed further in Section 3.2 of this chapter.

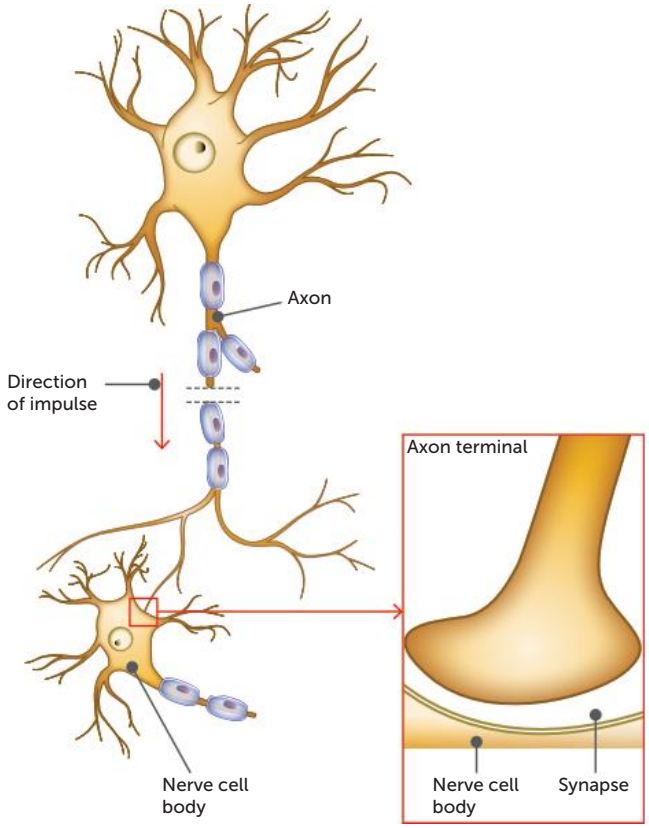

FIGURE 3.4 A synapse is a small gap between adjacent neurons

Types of neurons

Neurons can be classified based on their function or structure.

Functional types of neurons

Neurons may be classified into three types depending on the *function* each performs.

- **Sensory** (also known as **afferent** or **receptor**) **neurons** carry messages from receptors in the sense organs, or in the skin, to the central nervous system (brain and spinal cord).
- **Motor** (also known as **efferent** or **effector**) **neurons** carry messages from the central nervous system to the effectors, the muscles and glands.
- **Interneurons** are located in the central nervous system and are the link between the sensory and motor neurons. Interneurons may also be called **association neurons**, **connector neurons** or **relay neurons**.

Structural types of neurons

Another way of classifying neurons is by their *structure*. This classification is based on the number of extensions from the cell body.

- **Multipolar neurons** have one axon and multiple dendrites extending from the cell body. This type of neuron is the most common and includes most of the interneurons in the brain and spinal cord as well as the motor neurons that carry messages to the skeletal muscles.
- **Bipolar neurons** have one axon and one dendrite. Both the axon and dendrite may have many branches at their ends. Bipolar neurons occur in the eye, ear and nose, where they take impulses from the receptor cells to other neurons.
- **Unipolar neurons** have just one extension, an axon. These types of neurons are not found in humans or other vertebrates. They are found in insects.

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- **Pseudounipolar neurons** have properties of both unipolar neurons and bipolar neurons. There is a single axon from the cell body, which then separates into two extensions. One extension connects to dendrites, while the other ends in axon terminals. The arrangement of the cell body and axon means that the cell body lies to one side of the main axon. Most sensory neurons that carry messages to the spinal cord are of this type.

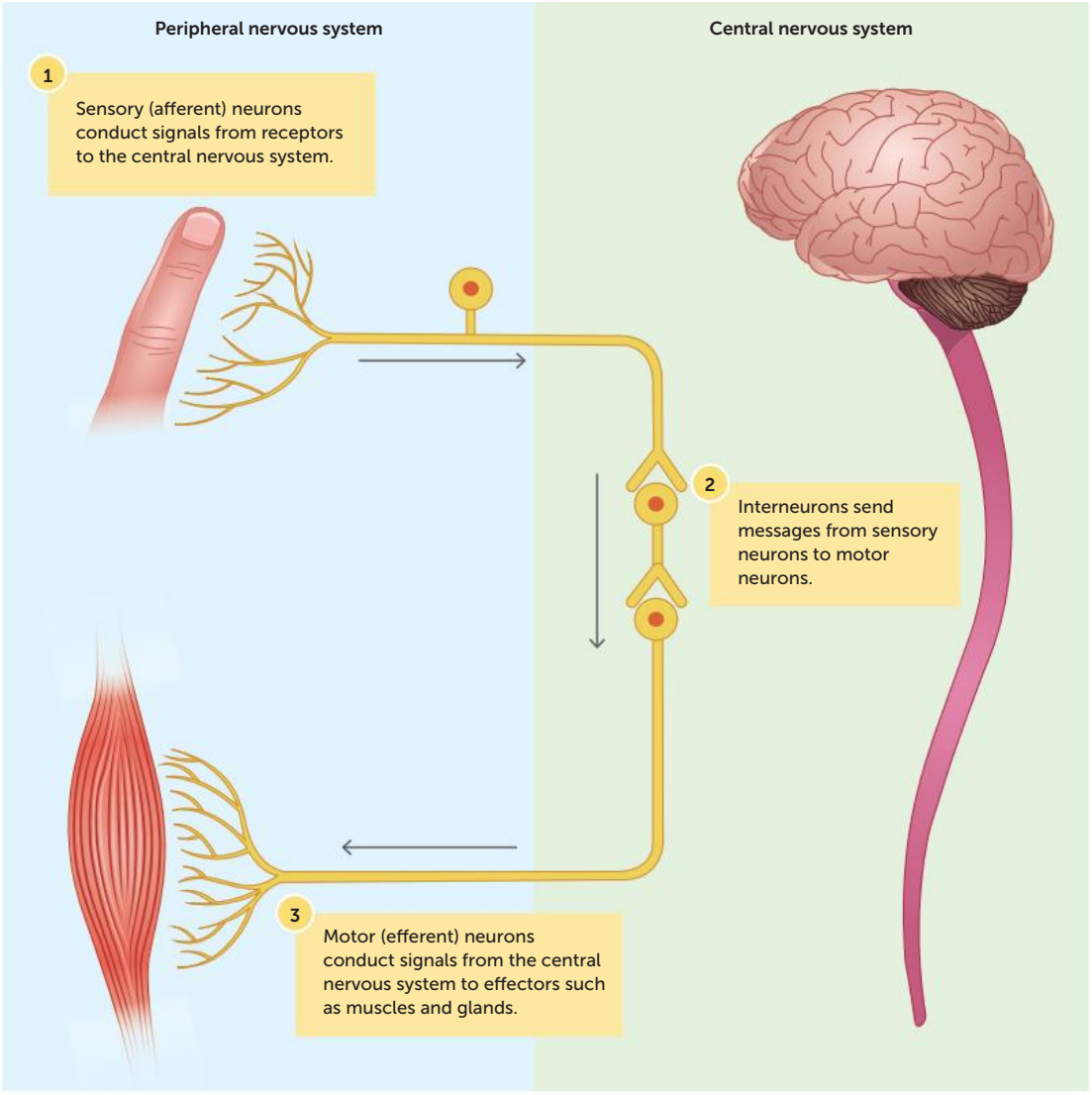

FIGURE 3.5 The direction of nerve impulses through sensory neurons, interneurons and motor neurons

TABLE 3.1 Summary of structural neurons

TYPE OF NEURON	NUMBER OF AXONS	NUMBER OF DENDRITES CONNECTING WITH THE CELL BODY	EXAMPLES OF NEURONS
Multipolar neurons	one	many	Motor neuron Interneuron
Bipolar neurons	one	one	Neurons in eye, ear and nose
Unipolar neurons	one	nil	Not found in humans
Pseudounipolar	one that divides into two	nil	Sensory neuron

WS
3.1 Neurons

Key concept

Neurons can be classified based on either their structure or their function.

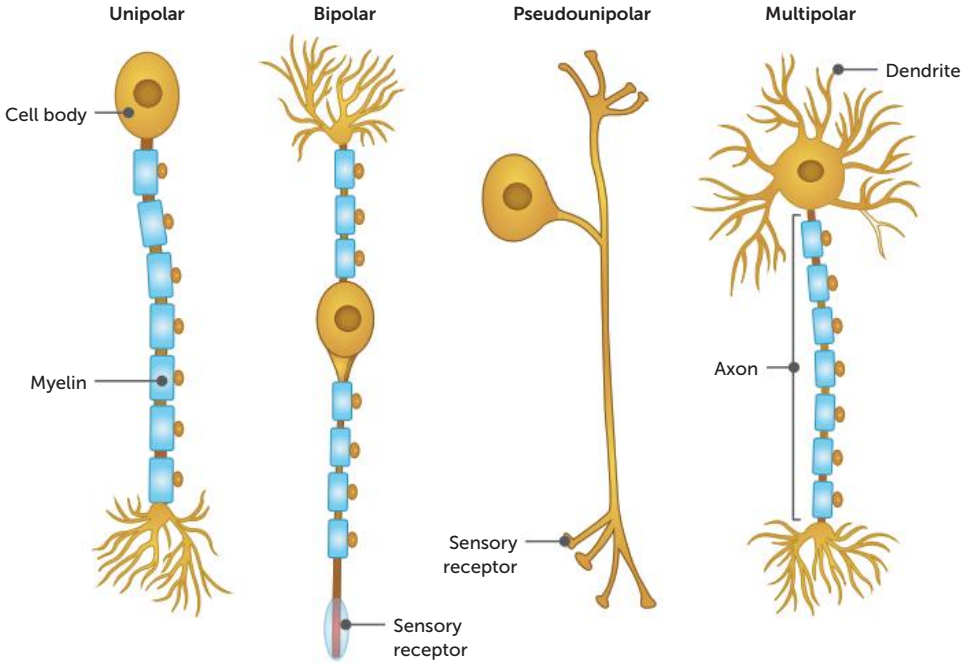

FIGURE 3.6 Types of neurons based on their structure

Nerve fibres

As previously mentioned, the axons and dendrites of nerve cells are known as nerve fibres. Outside the brain and spinal cord, nerve fibres are grouped together to form a **nerve**. Nerve fibres are arranged into bundles held together by connective tissue, with multiple bundles joining together to form a nerve.

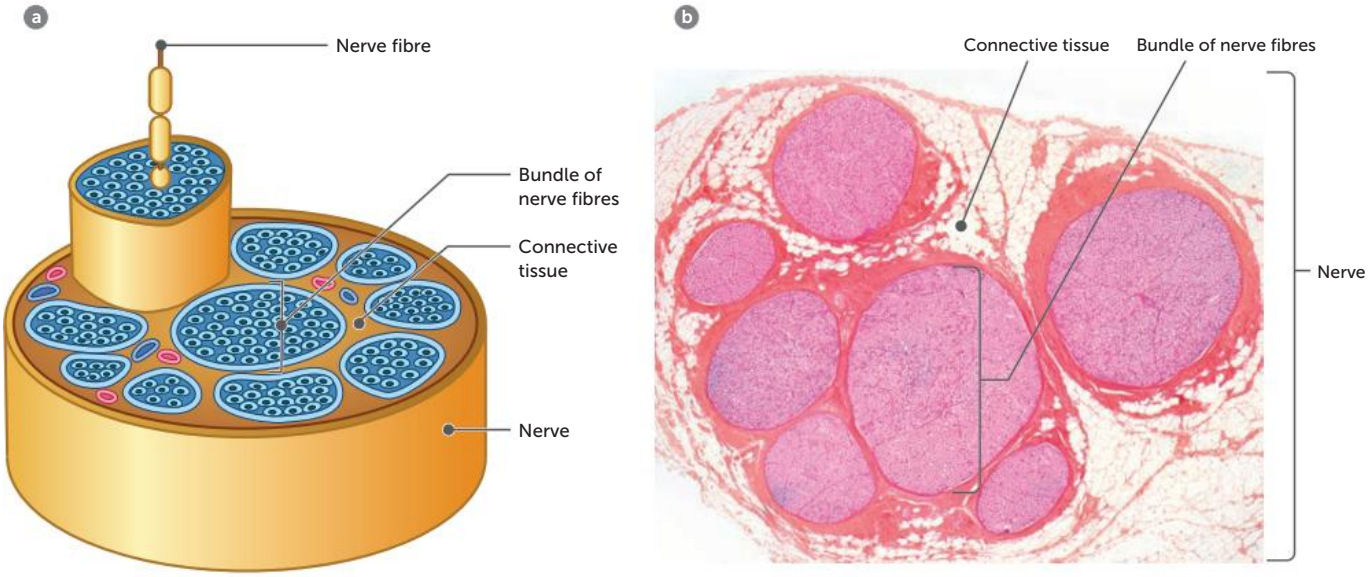

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FIGURE 3.7 a Cross-section of a nerve; **b** Cross-section of a nerve viewed under a microscope

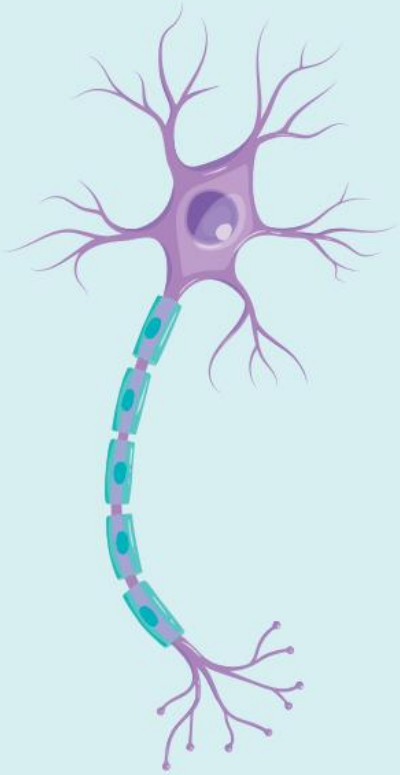
TABLE 3.2 The difference between neurons, nerve fibres and nerves

NEURON	A nerve cell
NERVE FIBRE	Any long extension of cytoplasm of a nerve cell body, although the term usually refers to an axon
NERVE	Bundles of nerve fibres held together by connective tissue

Questions 3.1

RECALL KNOWLEDGE

1 Label the diagram below to identify the axon, cell body, myelin sheath, dendrite, nucleus, axon terminal, cytoplasm, node of Ranvier and Schwann cell.



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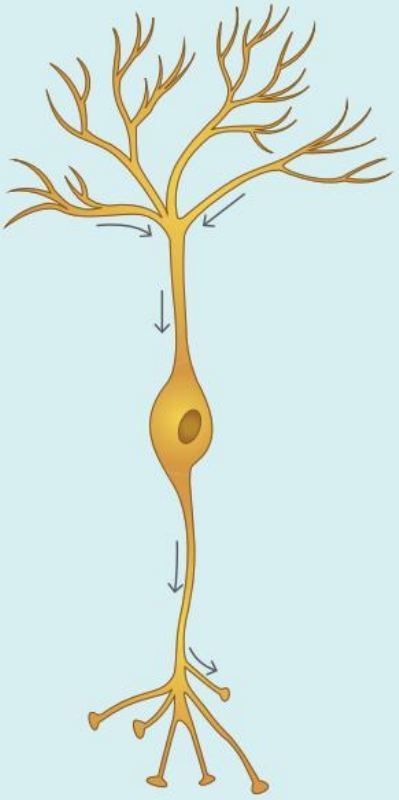
2 Complete the table to state the function of the parts of a neuron.

STRUCTURE	FUNCTION
Mitochondria	
Axon	
Dendrite	
Myelin sheath	
Neurilemma	

3 State the function of:

- **a** motor neurons
- **b** interneurons
- **c** sensory neurons.

4 Classify the neuron shown below and justify your choice.



5 Describe the arrangement of nerve fibres in a nerve.

6 Define 'synapse' and state its function.

APPLY KNOWLEDGE

7 Explain why the white matter in the brain is white in colour.

8 Compare and contrast a synapse and a neuromuscular junction.

9 Suggest why interneurons are multipolar in structure.

3.2 NERVE IMPULSES

The message that travels along a nerve fibre is called a **nerve impulse**. Nerve impulses are transmitted very quickly, making it possible for the body to respond rapidly to any change in the internal or external environment.

Conduction of a nerve impulse

A nerve impulse is an **electrochemical change** that travels along a nerve fibre. It is described as electrochemical because it involves:

- a change in *electrical* voltage
- that is brought about by changes in *chemicals* (specifically, the concentration of ions inside and outside the cell membrane of the neuron).

Electrical charge and potential difference

There are two types of electrical charges: positive and negative. Two positive or two negative charges *repel* each other. A positive and a negative charge *attract* each other. That is, like charges repel and opposite charges attract.

When opposite charges are separated, an electrical force tends to pull them together. The force that pulls unlike charges together can be measured, and its strength increases as the charges get closer or larger. When positive and negative charges come together, energy is released. If a group of positive and negative charges are separated, they have the potential to come together and release energy. The potential, or **potential difference**, between two places can be measured. It is called the voltage and is measured in volts (V) or millivolts (mV), where there are 1000 mV in 1 V.

Potential difference across a cell membrane

When some chemical substances are dissolved in water, they break up into electrically charged particles called **ions**. This happens to some of the substances dissolved in the fluid around and inside cells.

- The fluid outside the cell, the **extracellular fluid**, contains a high concentration of sodium chloride, and so most of its charged particles are positive sodium ions (Na^+) and negative chloride ions (Cl^-).
- The fluid inside the cell, the **intracellular fluid**, has a low concentration of sodium ions and chloride ions. Its main positive ions are potassium (K^+), and the negative ions come from a variety of organic substances made by the cell.

Differences in the concentration of ions mean that there is a potential between the inside and the outside of the cell membrane. This potential difference is called the **membrane potential**. It occurs in all body cells, but is particularly large in nerve and muscle cells. The membrane potential of unstimulated nerve cells, known as the **resting membrane potential**, can be measured and is about −70 mV. This means that the potential of the inside of the membrane is 70 mV less than that of the outside.

Key concept

The resting membrane potential of an unstimulated neuron is approximately −70 mV due to the fluid inside the cell being more negatively charged than the fluid outside of the cell.

Ions are unable to diffuse through the phospholipid bilayers of the cell membrane directly. Instead, they move through protein channels. Some channels, called **leakage channels**, are open all the time; others, called **voltage-gated channels**, only open when the nerve is stimulated.

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The resting membrane potential of neurons is due mainly to differences in the distribution of potassium ions (K^+) and sodium ions (Na^+) on either side of the cell membrane, making the extracellular fluid more positively charged than the intracellular fluid.

- The concentration of sodium ions is about 10 times higher outside the neuron than inside. The cell membrane is only slightly permeable to sodium ions due to the limited number of sodium leakage channels. This limits the facilitated diffusion of sodium ions.
- The concentration of potassium ions is about 30 times greater inside the neuron than outside. The cell is highly permeable to potassium due to the larger number of potassium leakage channels. Therefore, more potassium ions are able to diffuse than sodium ions.
- The concentration of chloride ions is higher outside the neuron than inside. The cell membrane is highly permeable to chloride ions, allowing their diffusion through protein channels.
- The concentration of large, negatively charged organic ions is higher inside the neuron than outside. The cell membrane is impermeable to these ions; therefore, they stay inside the cell.

In addition to protein channels, sodium and potassium ions move across the cell membrane through a carrier protein known as the **sodium–potassium pump**. The pump moves two potassium ions into the cell for every three sodium ions that are removed. Therefore, there is a net reduction of positive ions inside the cell. This movement is against the concentration gradient and, therefore, is active transport and uses adenosine triphosphate (ATP).

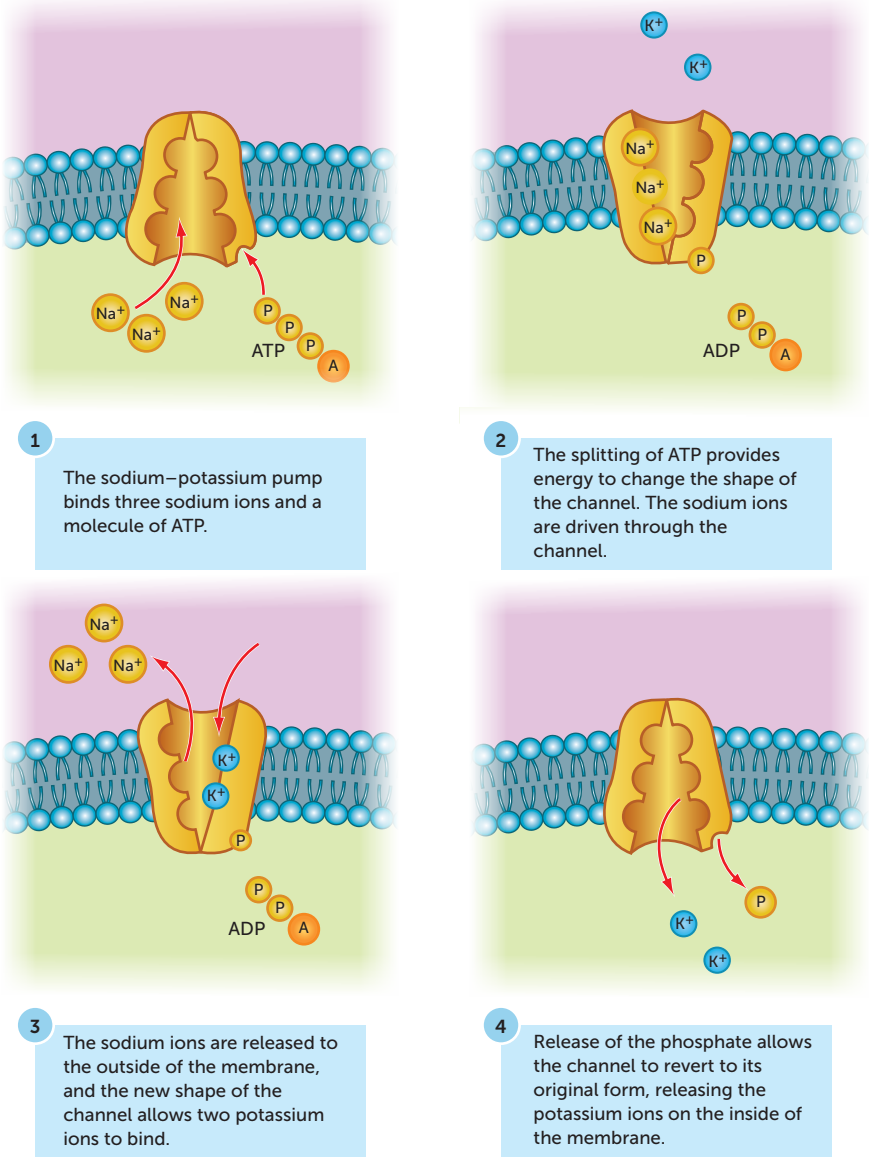

FIGURE 3.8 The sodium–potassium pump uses active transport to move three sodium ions out of the cell for every two potassium ions that move into the cell

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The combination of the location of the ions, the permeability of the cell membrane and the sodium–potassium pump means that there is a net flow of positive ions out of the cell because more potassium ions are diffusing out of the cell than there are sodium ions diffusing into the cell. This, in addition to the negative organic ions inside the cell, results in the inside of the cell being more negative than the outside. This produces a negative resting membrane potential, and the membrane is said to be **polarised**.

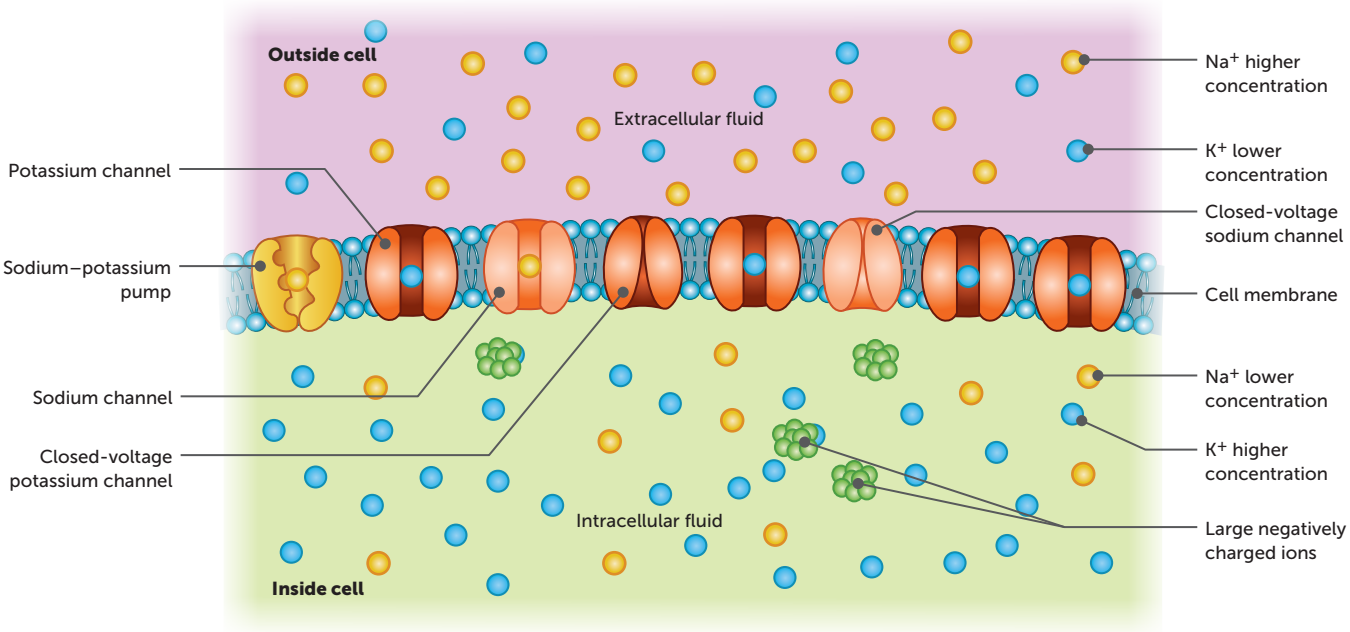

FIGURE 3.9 Relative concentrations of ions creating the resting membrane potential

Key concept

The resting membrane potential is maintained by a difference in the number of leakage channels for sodium and potassium ions, the membrane being impermeable to large organic negative ions and the sodium–potassium pump. This results in the intracellular fluid being less positively charged than the extracellular fluid.

Action potential

If the stimulus to a neuron is sufficient, the signal will be passed along the neuron. This happens due to the opening and closing of voltage-gated channels, which causes the rapid depolarisation and repolarisation of the membrane. This lasts approximately 1 millisecond and is called an **action potential**.

1 Depolarisation

Depolarisation is the sudden increase in membrane potential. This occurs if the level of stimulation exceeds about 15 mV, or the **threshold**.

When a neuron is stimulated by a neurotransmitter or a sensory receptor, some sodium channels are opened. These channels are known as ligand-gated or mechanical-gated channels. Once they are open, more sodium ions move into the cell. This makes the intracellular fluid less negative, increasing the potential difference.

If the stimulus is strong enough to increase the potential to −55 mV, then voltage-gated sodium channels open. This produces a movement of sodium ions into the cells that proceeds independently of the stimulus. That is, the size of the response is not related to the strength of the stimulus. This is known as an **all-or-none response**.

This inward movement of sodium ions is too great to be balanced by an outward movement of potassium ions, making the inside of the membrane more positive than the outside. The original polarity of the membrane increases, reaching approximately +40 mV. The membrane is then said to be **depolarised**.

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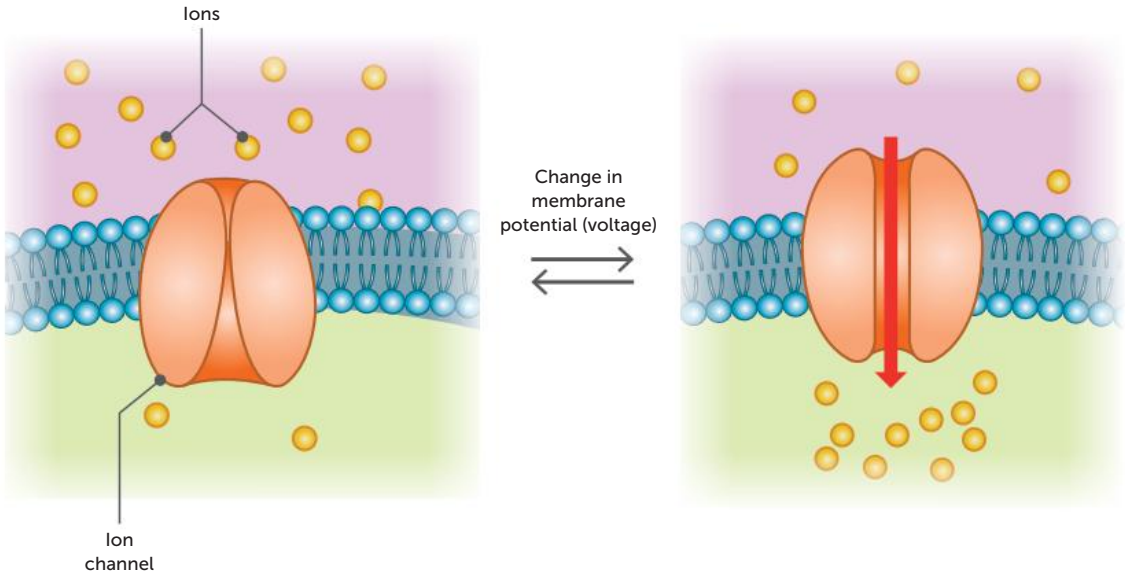


FIGURE 3.10 The opening of voltage-gated sodium ion channels leads to the movement of sodium ions into the cell, increasing the membrane potential

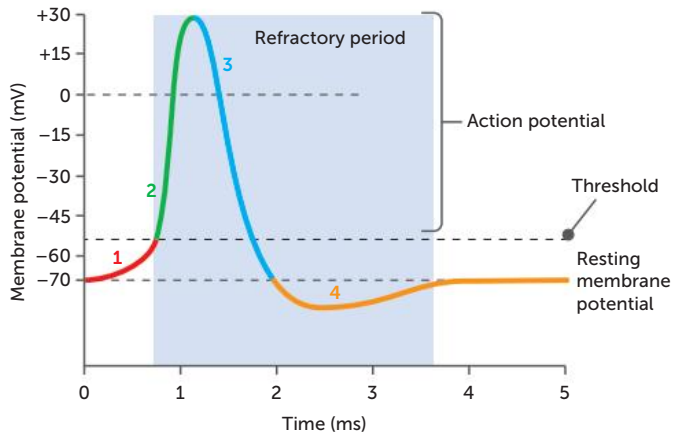
2 Repolarisation

After a short period, **repolarisation** occurs. The sodium channels close, which stops the influx of sodium ions. At the same time, voltage-gated potassium channels open, increasing the flow of potassium ions out of the cell. This makes the inside of the membrane more negative than the outside and decreases the membrane potential. The membrane is **repolarised**.

The potassium channels remain open longer than what is needed. This results in the membrane potential dropping lower than the resting membrane potential, and the membrane is **hyperpolarised**. This process is called **hyperpolarisation**.

3 Refractory period

Once the sodium channels have opened, they quickly become inactivated. This means that they are unresponsive to stimulus. Therefore, for a brief period after being stimulated, the membrane will not undergo another action potential. This period, called the **refractory period**, lasts from when the membrane reaches the threshold of −55mV until it returns to the resting membrane potential of −70mV.



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 is hyperpolarised and then returns to resting state.

FIGURE 3.11 Development of an action potential on a nerve cell membrane

3.2 Nerve impulses

Action potential
Watch a simulation of the changes in potential during an action potential.

Activity 3.2
Storyboarding an action potential

Key concept

An action potential is the rapid depolarisation and repolarisation of the membrane. A refractory period means that there is a period of time before another action potential can occur at the same location.

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Transmission of the nerve impulse

A single action potential occurs in one section of a membrane. However, it triggers an action potential in the adjacent membrane. This process continues along the length of the neuron and is called a nerve impulse.

Thus, an action potential does not travel along the nerve fibre; it is the message, or nerve impulse, that travels along the fibre. The situation has been likened to a line of dominoes. When the first domino falls it pushes over the second, which in falling pushes over the third, and so on. No one domino travels along the line, but the energy that triggers the fall is transmitted from the first domino to the last.

Conduction along unmyelinated fibres

In an unmyelinated nerve fibre, depolarisation of one area of the membrane causes a movement of sodium ions into the adjacent areas. This movement stimulates the opening of the voltage-gated sodium channels in the next part of the membrane, which initiates an action potential in that area of the membrane. The process repeats itself along the whole length of the membrane so that the action potential moves along the membrane away from the point of stimulation.

If the stimulus should occur in the middle of a fibre, impulses will travel in both directions along the fibre, away from the point of stimulation. However, in the body this would be unusual as stimulation normally occurs at the end of a fibre.

The nerve impulse is prevented from going backwards along the fibre by the refractory period. During the refractory period of an action potential, another action potential cannot be generated at that point on the fibre and so the nerve impulse is unable to travel in that direction.

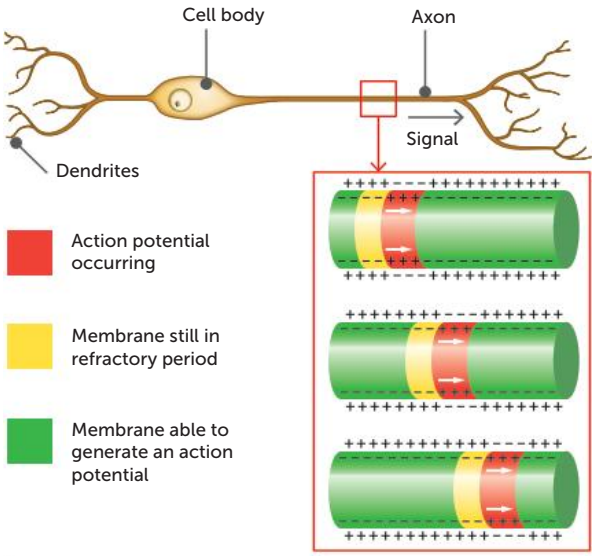

FIGURE 3.12 Transmission of a nerve impulse along an unmyelinated fibre. Successive action potentials are generated along the membrane of the nerve fibre

Transmission along myelinated fibres

In a myelinated fibre, the myelin sheath insulates the nerve fibre from the extracellular fluid. This does not occur at the nodes of Ranvier because the myelin sheath is absent from the nodes. Therefore, where the nerve fibre is surrounded by myelin, ions cannot flow between the inside and outside of the membrane and an action potential cannot form. Instead, the action potential jumps from one node of Ranvier to the next. This 'jumping conduction', known as **saltatory conduction**, allows the nerve impulse to travel much faster along myelinated fibres than along unmyelinated ones. A large myelinated fibre can conduct impulses at a speed of up to 140 metres per second (m/s); in an unmyelinated fibre the maximum speed of transmission is 2 m/s.

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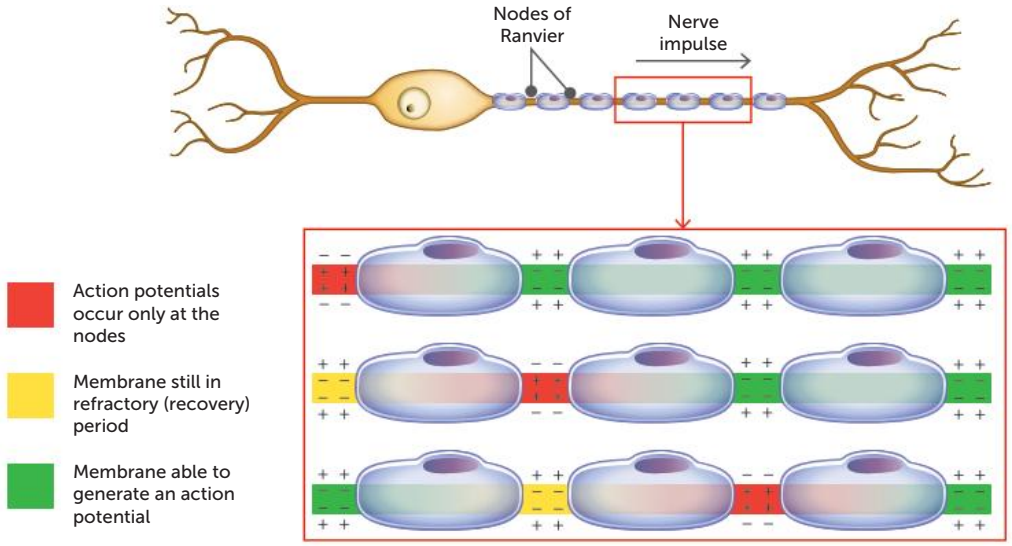

FIGURE 3.13
Saltatory conduction along a myelinated fibre. The nerve impulse jumps from node to node

Key concept

Nerve impulses travel much faster along a myelinated fibre due to saltatory conduction.

Size of the nerve impulse

A nerve impulse that travels along a fibre is always the same size, regardless of the size of the stimulus. A weak stimulus, provided it exceeds the threshold, produces the same action potential as a strong one. As mentioned before, this is called an all-or-none response – a stimulus is either strong enough to trigger an impulse, or it is not. The magnitude of the impulse is always the same. If you stub your toe, nerve impulses will be generated that travel along an axon all the way up to your spinal cord. The voltage of the impulses arriving at the spinal cord will be the same as the voltage of those generated at the toe. This situation has been likened to a burning fuse. When the fuse is lit, the heat generated ignites the next part of the fuse, which then produces enough heat to light the next part, and so on. The end of the fuse burns with the same amount of heat as the beginning. Like the heat in a burning fuse, a nerve impulse does not become weaker with distance.

How is it, then, that we are able to distinguish stimuli of different intensities? For example, how do we tell a light tap on the shoulder from a heavy slap on the back? Two things enable us to determine the strength of a stimulus: a strong stimulus causes depolarisation of more nerve fibres than a weak stimulus; and a strong stimulus produces more nerve impulses in a given time than a weak stimulus.

Transmission across a synapse

The synapse is the very small gap between adjacent neurons. We have seen how the nerve impulse is transmitted along the membrane of a neuron by a change in the ion concentration on each side of the membrane, but at the synapse there is no membrane and so some other method of transmission must be involved. The process is as follows:

1. When the nerve impulse reaches the axon terminal, it activates voltage-gated calcium ion channels.
2. As there is a higher concentration of calcium ions in the extracellular fluid, they flow into the cell at the pre-synaptic axon terminal.

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Activity 3.3
Examining the discovery of neurotransmitters

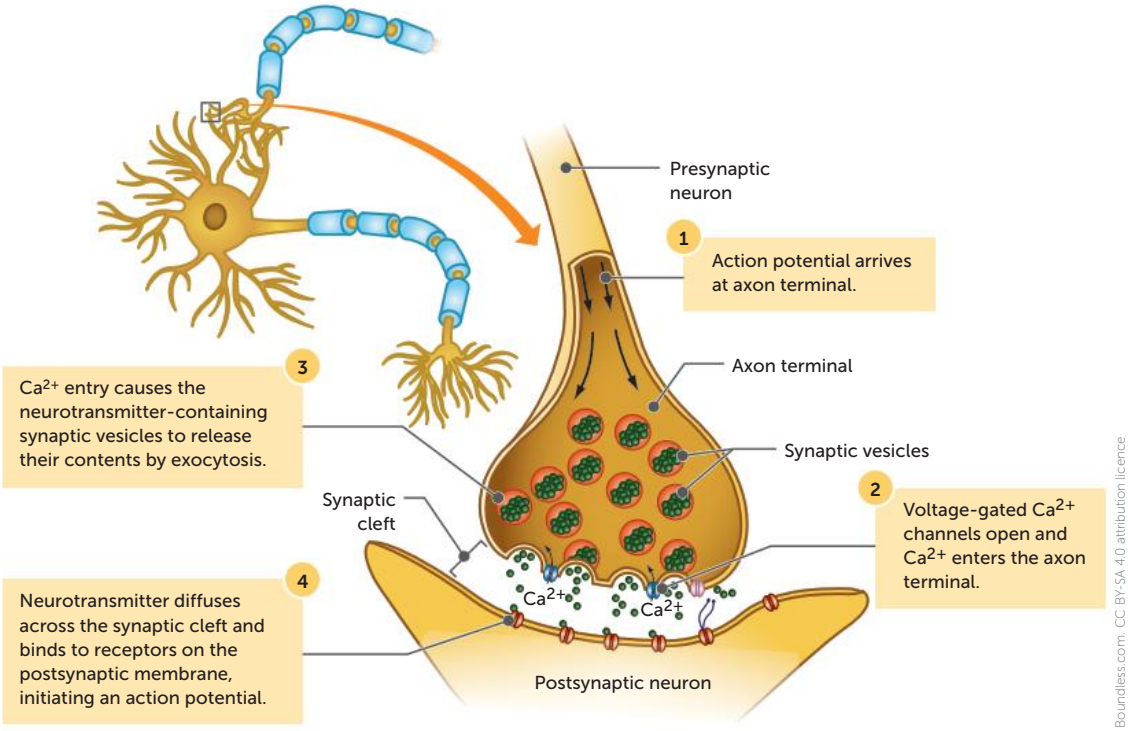
3 This causes synaptic vesicles to fuse with the membrane, releasing special chemicals called neurotransmitters by exocytosis.
4 The neurotransmitters diffuse across the gap and attach to receptors on the membrane of the next neuron.
5 This stimulates ligand-gated protein channels to open, which allows the influx of sodium ions and initiates an action potential in the post-synaptic membrane.

Neurotransmitters are removed from the synapse by being reabsorbed by the presynaptic membrane, by being degraded by enzymes or by moving away through diffusion.

More than 100 different substances are either confirmed as or suspected of being neurotransmitters, including acetylcholine, adrenaline, dopamine and histamine.

The transmission of nerve impulses across a synapse occurs in only one direction – from axon to dendrite or from axon to cell body.

FIGURE 3.14
Transmission of a nerve impulse across a synapse



Chocolate and neurotransmitters
Do you like chocolate? This website explores how chocolate may have an effect on neurotransmitters in the brain.

Nerve agents
This website provides more information about nerve agents.

Key concept

Nerve impulses travel from one neuron to the next by neurotransmitters diffusing across the synapse.

Effect of chemicals on the transmission of nerve impulses

There are many chemicals, both natural and synthetic, that influence the transmission of nerve impulses, mostly at the synapse or at the neuromuscular junction. Stimulants such as caffeine and benzedrine stimulate transmission at the synapse. Other drugs, such as anaesthetics or hypnotics, depress the transmission. Venom from certain species of snakes and spiders also affects the neuromuscular junction.

Nerve agents (also called nerve gases) contain organophosphates, which cause the build-up of acetylcholine at the neuromuscular junction. All muscles in the body then try to contract and the loss of muscle control prevents breathing. Organophosphates are also used in some insecticides.

Activity 3.4
Investigating synapse response in *Daphnia*

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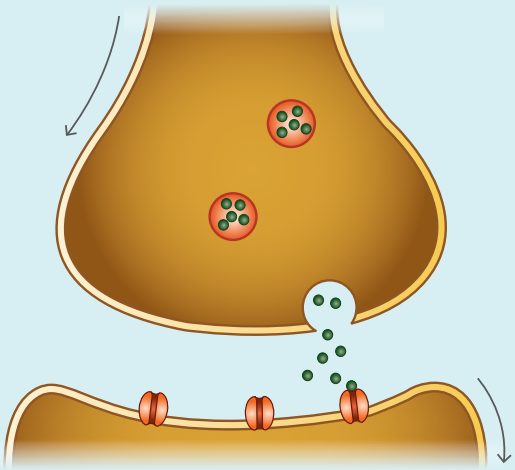
Questions 3.2

RECALL KNOWLEDGE

1 Describe the difference between an action potential and a nerve impulse.
2 Are there more sodium leakage channels or potassium leakage channels in the membrane of a neuron?
3 Explain the role of large organic ions in establishing the resting membrane potential.
4 Use a flow chart to describe the events that happen during an action potential.
5 Describe the concept of an all-or-none response in relation to an action potential and the relevance of the threshold.
6 Describe how a nerve impulse travels along a nerve fibre.
7 Draw, and label, a graph representing the membrane potential before, during and after an action potential.
8 List the ways in which a large stimulus is able to be recognised as different from a weak stimulus.
9 Add labels and additional structures to the diagram below to show how a nerve impulse travels from one neuron to the next.
10 Describe the process of saltatory conduction.

APPLY KNOWLEDGE

11 Explain the difference between a membrane potential of –70 mV and +40 mV.
12 Suggest how the resting membrane potential would be different if the sodium–potassium pump did not work.
13 Compare and contrast the progression of a nerve impulse along a myelinated fibre and along an unmyelinated fibre.
14 Explain why organophosphate poisoning results in continual muscle contractions.



3.3 RECEPTORS AND REFLEXES

A **receptor** is a structure that is able to detect a change in the body's internal or external environment. Sometimes receptor cells of a particular type are grouped together in a **sense organ**, such as the light receptors in the eye or the receptors sensitive to sound vibrations in the ear. Other receptors are simple nerve endings and may be spread through parts of the body or even the whole body, such as pain receptors or the temperature receptors in the skin. When a receptor is stimulated, the body is able to respond to the change. In some cases, this is via an automatic reflex; in other cases the response is more complex.

Types of receptors

Changes in the environment may come from different sources. Therefore, there are different types of receptors to be able to detect the different types of **stimuli**.

Thermoreceptors

Thermoreceptors are able to respond to heat and cold. Skin thermoreceptors inform the brain (the hypothalamus and the cerebrum) of changes in the temperature outside the body. In this way, we are consciously aware of the temperature of our surroundings. Peripheral thermoreceptors in the skin are nerve endings that are sensitive to either heat or cold, but not both. If the skin is tested with hot and cold probes, it is found that there are definite hot spots and cold spots, depending on the type of thermoreceptors present.

The temperature inside the body, the **core temperature**, is monitored by thermoreceptors in the hypothalamus, which detect the temperature of the blood that is flowing through the brain. Using information received from the skin and from its own thermoreceptors, the hypothalamus can regulate body temperature. This process will be discussed further in Chapter 5.

Osmoreceptors

Osmotic pressure is determined by the concentration of substances dissolved in the water of the blood plasma. The higher the concentration, the greater the osmotic pressure. **Osmoreceptors** are located in the hypothalamus and are sensitive to even very small changes in osmotic pressure. They can stimulate the hypothalamus so that the body's water content is maintained within very narrow limits. This process will be discussed further in Chapter 6.

Chemoreceptors

Chemoreceptors are stimulated by particular chemicals. They are present in the nose, making us sensitive to odours, and in the mouth, giving us sensitivity to tastes. There are also internal chemoreceptors that are sensitive to the composition of body fluids. Of particular importance are chemoreceptors in certain blood vessels that are sensitive to the pH of the blood and to the concentrations of oxygen and carbon dioxide. These chemoreceptors are involved in the regulation of the heartbeat and of breathing, which will be discussed in Chapter 6.

Touch receptors

Touch receptors (also known as mechanoreceptors or pressure receptors) are found mainly in the skin. There are a number of different types of touch receptors. Some are close to the surface of the skin and are sensitive to very light touch. These occur in greater concentrations in areas such as the lips, fingertips, eyelids and external genital organs.

Nerve endings are also associated with the base of each hair follicle. These respond to any light touch that bends the hair. Touch receptors close to the skin surface and those attached to the hairs adapt rapidly, and so after a short time we are no longer aware of the touch. For example, when first putting on clothing we are aware of it touching the skin, but that sensation disappears very quickly.

Other touch receptors are located deeper in the skin and are sensitive to pressure and vibrations.

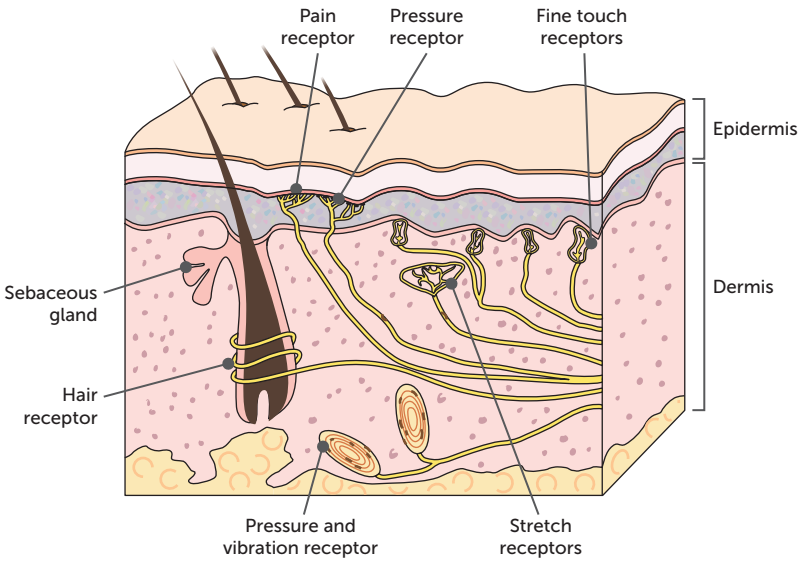


FIGURE 3.15 Section of the skin showing receptors

Pain receptors

Pain receptors (also called nociceptors) are stimulated by damage to the tissues, such as from a cut or a heavy bump, by poor blood flow to a tissue, or by excessive stimulation from stimuli such as heat or chemicals. The receptors for pain are especially concentrated in the skin and the mucous membranes. They occur in most organs, but not in the brain.

Pain is uncomfortable, but it is essential for our wellbeing. Pain warns us that damage to tissues is occurring, and we can therefore take evasive action or seek medical help so that damage is minimised.

Unlike many other receptors, pain receptors adapt little or not at all, so that pain continues for as long as the stimulus is present. In some cases, prolonged stimulation of pain receptors makes the pain worse. The failure of pain receptors to adapt keeps the person aware that a tissue-damaging situation exists.

Reflexes

A **reflex** is a rapid, automatic response to a change in the external or internal environment. All reflexes have four important properties.

- A *stimulus* is required to trigger a reflex – the reflex is not spontaneous.
- A reflex is *involuntary* – it occurs without any conscious thought.
- A reflex response is *rapid* – only a small number of neurons are involved.
- A reflex response is *stereotyped* – it occurs in the same way each time it happens.

Some reflexes involve the unconscious parts of the brain, but most are coordinated by the spinal cord. When a nerve impulse comes into the spinal cord from a receptor, the message is not necessarily carried up to the brain. The impulse may be passed to motor neurons at the same level in the cord, or it may travel a few segments up or down the cord before travelling out through a motor neuron. In these cases, the reflex is carried out by the spinal cord alone and is known as a **spinal reflex**. The pathway a nerve impulse follows in travelling from a receptor to an effector is known as a **reflex arc** or, in the case of a spinal reflex, a **spinal reflex arc**.

Even though contraction of skeletal muscles may occur in a spinal reflex, it does not involve the brain and therefore is involuntary. Impulses may be sent to the brain, and so we become aware of what is happening, but this awareness does not occur until after the response has been initiated. For example, if you step on something sharp with bare feet, the reflex response is to withdraw your foot from the painful stimulus. By the time your brain becomes consciously aware of the painful stimulus, your foot has already been withdrawn.

A reflex arc has the following basic components.

- The *receptor* reacts to a change in the internal or external environment by initiating a nerve impulse in the sensory neuron.
- A *sensory neuron* carries impulses from the receptor to the spinal cord or brain.
- There is at least one *synapse*; the nerve impulse may be passed directly to a motor neuron, or there may be one or more interneurons that direct the impulse to the correct motor neuron.
- A *motor neuron* carries the nerve impulse to an effector.
- An *effector* receives the nerve impulse and carries out the appropriate response. Effectors are muscle cells or secretory cells.

Figure 3.16 shows the components in a simple spinal reflex involving three neurons. The response would occur in a fraction of a second; while it was occurring, impulses would travel up the spinal cord to the brain. Only after the response had been made would the person become consciously aware of the situation. Many reflexes, such as withdrawing the hand from a painful stimulus, protect the body from injury. Blinking when something touches the cornea of the eye, sneezing or coughing when something irritates the nose or trachea, and constriction of the pupil in response to intense light are other protective reflexes.

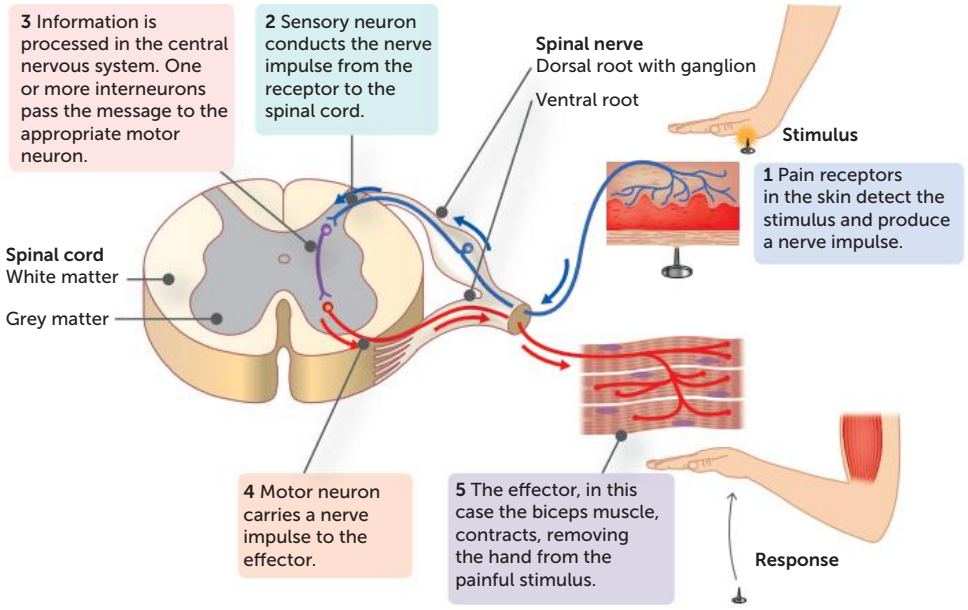


FIGURE 3.16 The neurons involved and the pathway followed by the nerve impulses in a spinal reflex. In this example, the impulses enter and leave the spinal cord by the same spinal nerve. This is not always the case

Simple reflexes and reflex arcs
This website provides more information about simple reflexes and reflex arcs and an animation of the latter.

Other reflexes include secretion of saliva in response to the sight, smell or taste of food, the ejaculation of semen during sexual intercourse, and the responses brought about by the autonomic nervous system.

Learnt reflexes

The protective reflexes mentioned above are present from birth. More complex motor patterns appear during a baby's development, including reflexes such as suckling, chewing or following movements with the eyes. These **innate reflexes** are determined genetically.

Some complex motor patterns are learnt and are called **acquired reflexes**. Muscular adjustments required to maintain balance while riding a bike, jamming on the brakes of a car to avoid a dangerous situation, or catching a ball are all acquired reflexes. They are learnt through constant repetition.

Activity 3.5
Investigating reflexes

Activity 3.6
Investigating reaction time

Activity 3.7
Testing more reaction times

Questions 3.3

RECALL KNOWLEDGE

1. List the different types of receptors and state the relevant stimulus for each.
2. List the properties of all reflexes.
3. Draw a labelled diagram to represent a spinal reflex arc.

APPLY KNOWLEDGE

4. Compare and contrast pain and touch receptors.
5. Explain how a gag reflex protects the body.
6. Describe the steps involved in the reflex initiated by touching a hot object.

3.4 COMPARISON OF HORMONAL AND NERVOUS COORDINATION

Both the endocrine system and the nervous system are involved in communication within the body. However, they do not duplicate each other's roles; rather, they complement and reinforce each other.

The differences between the actions of nerves and hormones are as follows.

- Nervous responses are more rapid than hormonal ones, because nerve impulses travel rapidly along nerve fibres, while hormones are transported in the bloodstream. The nervous system

responds to a stimulus in milliseconds, while the release of hormones may take from several seconds to several days.

- When a stimulus ceases, the nervous system stops generating nerve impulses and the response ceases almost immediately. Thus, nerve impulses bring about an immediate response, which lasts for only a short time. Hormones are typically slower acting, and responses can last a considerable time (even for years).
- Nervous messages are an electrochemical change that travels along the membrane of a neuron. Endocrine messages are chemicals (hormones) that are usually transported by the blood.
- Nerve impulses travel along a nerve fibre to a specific part of the body and often influence just one effector; hormones travel to all parts of the body, are carried by the blood and often affect a number of different organs.

It should be stressed that these differences are only generalisations. There are exceptions to each of them. For example, response to the hormone adrenaline can be quite rapid, and some chemical messengers are not carried by the blood because their site of action is adjacent to the cells in which they are produced.

Despite these differences there are important overlaps between the two systems.

- Some substances function as both hormones and neurotransmitters. Examples are noradrenaline, antidiuretic hormone and dopamine.
- Some hormones such as oxytocin and adrenaline are secreted by neurons into the extracellular fluid.
- Some hormones and neurotransmitters have the same effect on the same target cells. For example, noradrenaline and the hormone glucagon both act on liver cells to cause glycogen to be broken down into glucose.

Table 3.3 compares the nervous and endocrine systems.

TABLE 3.3 A comparison of the nervous and endocrine systems

CHARACTERISTIC	NERVOUS SYSTEM	ENDOCRINE SYSTEM
Nature of message	Electrical impulses and neurotransmitters	Hormones
Transport of message	Along the membrane of neurons	By the bloodstream
Cells affected	Muscle and gland cells; other neurons	All body cells
Type of response	Usually local and specific	May be very general and widespread
Time taken to respond	Rapid – within milliseconds	Slower – from seconds to days
Duration of response	Brief – stops quickly when the stimulus stops	Longer lasting – may continue long after the stimulus has stopped

Questions 3.4

RECALL KNOWLEDGE

1 For each property listed below, state whether it describes the nervous system or the endocrine system.
   - **a** Has a specific target.
   - **b** Produces a long-lasting effect.
   - **c** Message is carried through the bloodstream.
   - **d** Is slow to respond to a stimulus.
   - **e** Messages travel due to an electrochemical change.
   - **f** Affects muscles, glands and other neurons.
   - **g** Is quick to respond.
   - **h** Affects all body cells.
   - **i** Effects last a short time.

2 State two ways in which the nervous system is similar to the endocrine system.

APPLY KNOWLEDGE

3 Explain why the body needs both the endocrine and nervous systems.

4 Suggest which system (nervous or endocrine) would have the biggest effect on heart rate. Justify your answer.

CHAPTER 3 ACTIVITIES

ACTIVITY 3.1 Creating a model of a neuron

Models are a useful way to represent information. In this activity, you will create a model of a multipolar neuron with a myelinated axon.

1. In groups of two or three, brainstorm the various components that you will need to show in your model and the materials you will represent them with. For example, you might use play dough, plasticine, lollies and pipe cleaners.
2. Create your model.
3. Take a photo of your model.
4. Annotate or label all the structures that make up your model of a neuron.
5. Make a video of your model by using your photo and adding a voice-over to explain the structures and their functions.

ACTIVITY 3.2 Storyboarding an action potential

Imagine that you are a sodium ion in the extracellular fluid around a neuron. Tell the story of your life during times when the neuron is unstimulated as well as when the neuron is sending a message in the body.

To make your story as accurate as possible, brainstorm the following:

1. What will be the setting of your story?
2. What will you (the ion) be doing during each of the different situations?
3. What other characters will be in your story?
4. What is the plot of the story?

Write your story using clear explanations. Include relevant scientific terms to ensure the accuracy of the information.

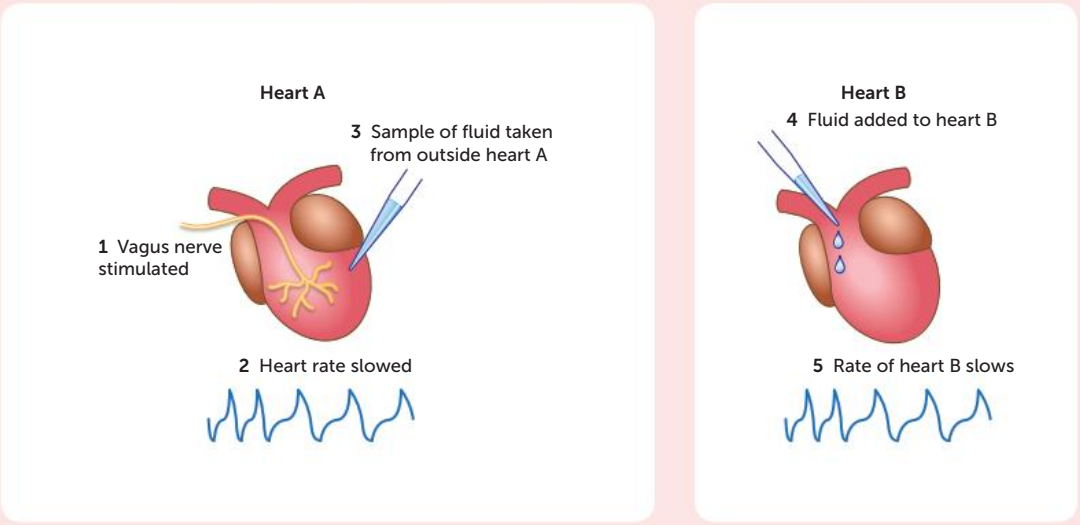
ACTIVITY 3.3 Examining the discovery of neurotransmitters

Neurotransmitters are chemicals released from a neuron that cause a response in an adjacent neuron, muscle or organ. A German pharmacologist, Otto Loewi, was the first person to demonstrate that nerve impulses exert their effect on muscles through the secretion of a neurotransmitter. In 1921 he performed an experiment that showed conclusively the effect of neurotransmitters. The idea for Loewi's experiment came to him in a dream. In his book *From the Workshop of Discoveries* (1953, University of Kansas Press), he said:

> *In the night of Easter Saturday, 1921, I awoke, turned on the light, and jotted down a few notes on a tiny slip of paper. Then I fell asleep again. It occurred to me at six o'clock in the morning that during the night I had written down something most important, but I was unable to decipher the scrawl. That Sunday was the most desperate day in my whole scientific life. During the next night, however, I awoke again, at three o'clock, and I remembered what it was. This time I did not take any risk; I got up immediately, went to the laboratory, made the experiment on the frog's heart, described above, and at five o'clock the chemical transmission of nervous impulse was conclusively proved.*

The experiment that Loewi performed involved the use of two hearts from freshly killed frogs. The still beating hearts were placed in separate beakers of salt solution. Heart A still had the vagus nerve attached; heart B did not. When the vagus nerve of heart A was electrically stimulated, the heart slowed down. Loewi then took a dropper of the salt solution from around the slowly beating heart A and placed the fluid into the salt solution surrounding heart B. After a short time, heart B slowed down. Loewi concluded that a chemical produced by the vagus

nerve of heart A had caused heart B to slow down. In 1936, Loewi was awarded a Nobel Prize for his discovery.



Questions

1. Explain how the result of Loewi's experiment enabled him to claim that a chemical was involved in slowing the rate of beating of the hearts.
2. Would Loewi have gotten the same result if he had placed both hearts in the same beaker of salt solution?
3. What control experiments would have been necessary before Loewi could claim that a chemical secreted by nerve cells was involved in slowing the hearts?
4. Loewi called the chemical 'vagusstoff' (or 'vagus stuff' when translated into English). Find out what we now call the neurotransmitter that is released at neuromuscular junctions.
5. If Loewi was doing such an experiment today, what do you think he would write down as his:
   a. hypothesis?
   b. prediction?

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ACTIVITY 3.4 Investigating synapse response in *Daphnia*

Daphnia, commonly known as 'water fleas', are small freshwater invertebrate animals with an exoskeleton and paired appendages. *Daphnia* are translucent, making them an excellent organism in which to observe digestion and to study metabolic rates. This also makes *Daphnia* a great organism for studying homeostasis, as its clear external skeleton (carapace) allows visibility of the heart, located in its back. Homeostasis is the maintenance of a stable internal environment. Homeostatic mechanisms within animals are triggered by increases in cellular respiration. These mechanisms increase breathing and heart rate, so that more oxygen is available to cells and more carbon dioxide is removed from cells.

Aim

To investigate how different stimulants and depressants affect the neurotransmitters within *Daphnia*

Time requirement: 45 minutes

You will need

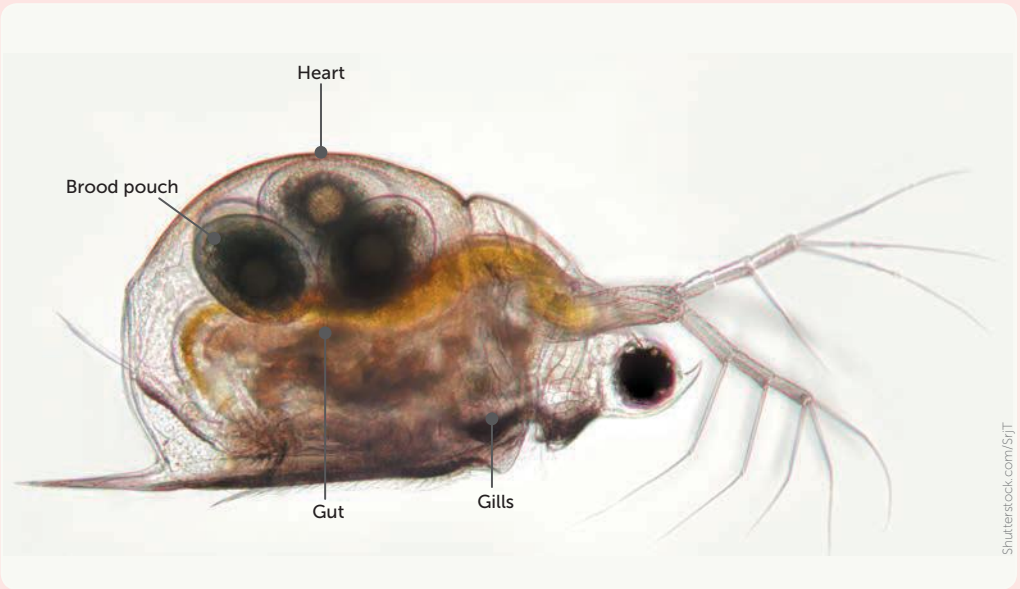
Concavity slides; 1% ethanol solution (1 drop); 1% caffeine solution (1 drop); 0.01% nicotine solution (1 drop); 1 plastic pipette (with the end cut); 3 plastic pipettes; *Daphnia* in culture; specimen container; cotton wool; vial for *Daphnia* disposal; paper towels; clock or stopwatch; compound microscope; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Daphnia are harmless to humans, but swamp or pond water may contain pathogens.	Wash hands after touching *Daphnia*.
Ethanol is highly flammable.	Store and use away from ignition sources. Do not heat in a container over an open flame; use a water bath that is spark-proof.
Nicotine may cause eye irritation.	Wear lab coats, safety glasses and gloves; wash hands thoroughly at the end.
Caffeine can be toxic at high doses.	Do not consume.

What to do

1. Place a very small piece of cotton wool in the centre of a concavity slide.
2. Using a plastic pipette with the end cut off, carefully transfer a *Daphnia* along with one drop of the culture liquid on to the slide (on top of the cotton). Keep the liquid to a minimum to prevent the *Daphnia* from swimming out of your field of view. If necessary, use a paper towel to draw off some water. Take care to leave some water for the *Daphnia*, and do not touch it with the paper towel because it will stick to the paper.
3. Place the slide under the compound microscope and adjust the focus until the *Daphnia* is in clear view. You should be able to clearly see the beating of the heart. Use the image below as a guide to locate the heart.



4. Ensure the microscope light is turned off to avoid overheating the *Daphnia*.

Measuring control heart rate

5 Using a stopwatch, count the number of heartbeats you observe in 10 seconds. You may wish to do this in pairs, so you can count the heartbeats as your partner keeps time. Try to take your measurements as quickly as possible, as the *Daphnia* will become stressed when kept in a small volume of water for an extended period.
6 Fill in the number of heartbeats in 10 seconds in the table below, and multiply the number by six to find the number of beats per minute.
7 Repeat this test twice until you have three separate heart rate measurements, then carefully transfer the *Daphnia* into the used-*Daphnia* vial. Calculate the average heart rate of the three measurements. This will serve as your control.

Measuring heart rate under the influence of chemicals

8 Using a new plastic pipette, add one drop of 1% caffeine solution to a new concavity slide.
9 Transfer a single *Daphnia* on to it, using the same technique as previously. Try to add as little culture liquid as possible when transferring the *Daphnia* to avoid diluting the caffeine too much.
10 Wait 30 seconds, then turn on the microscope light and count the number of heartbeats in 10 seconds. Record the result in the table.
11 Repeat this test twice so that you have three separate heart rate measurements. Multiply each number by six to generate the heart rate per minute. Calculate the average of your three measurements.
12 Carefully discard the *Daphnia* into the used-*Daphnia* vial, and repeat steps 8 to 11 for nicotine and ethanol using a fresh *Daphnia* and slide each time. Record your results in the table.

Studying your results

1 Copy and complete the table below with the results of your experiment.

	CHEMICALS							
	CONTROL		STIMULANTS				DEPRESSANT	
			CAFFEINE		NICOTINE		ALCOHOL (ETHANOL)	
TRIAL	10 sec.	BPM (x6)	10 sec.	BPM (x6)	10 sec.	BPM (x6)	10 sec.	BPM (x6)
1								
2								
3								
AVERAGE								

2 Which chemical caused the *Daphnia* heartbeat to beat the fastest?
3 Which chemical caused the *Daphnia* heartbeat to beat the slowest?

Discussion

1 Evaluate the accuracy of your counting method. Suggest how the accuracy of the procedure might be improved.
2 Describe how each of these chemicals affects the heart rate of the *Daphnia*.
3 There are two kinds of neurotransmitters: inhibitory and excitatory. Excitatory neurotransmitters stimulate the brain. Inhibitory neurotransmitters calm the brain and help create balance. Describe how the chemicals you tested affect neurotransmitters in the *Daphnia*.

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4 What factors can cause neurotransmitter levels to become out of balance? Describe how imbalances in neurotransmitter levels may affect human health.

Taking it further

Test how temperature changes the heart rate in *Daphnia*.

ACTIVITY 3.5 Investigating reflexes

In this activity, you will examine some simple reflex responses.

You will need (for each pair)

Ruler

What to do

For each of the following tests, one member of the pair should act as the subject, the other as the investigator. After you have conducted the tests once, swap roles and test the reflexes of the other person.

Knee reflex

The subject should sit on a stool or a bench-top with one leg crossed over the other. Using a ruler, the investigator should lightly strike the subject's crossed leg just below the kneecap.

1 Describe the response that occurs. The stimulus for the response is the stretching of the patellar tendon just below the kneecap.
2 Describe the location of the muscle or muscles that produce the response.
3 Describe, in words, the reflex arc that is involved in the response. Try to get a response with the knee straight and bent at different angles.
4 Does the response seem to be stronger at any particular angle of flexion? If so, can you suggest an explanation?

Heel reflex

Stand the subject beside a stool or chair with one leg kneeling on the seat of the chair. Use a ruler to strike the back of the subject's ankle.

5 Describe the response.
6 What is the stimulus in this case?
7 In what ways is the heel reflex similar to the knee reflex?
8 Doctors often test reflexes such as the knee and heel reflex. What do you think testing such reflexes would tell a doctor?

Eye reflex

With the subject seated directly in front, the investigator should suddenly clap their hands in front of the subject's face.

9 Describe any response observed.
10 Is the response a natural or a learnt response?
11 Does the response have a purpose? Explain.

Conclusions

1 Do all the reflexes that you have investigated have the four important properties that were described in this chapter?
2 Write a brief statement summarising the importance of reflexes to the normal functioning of the human body.

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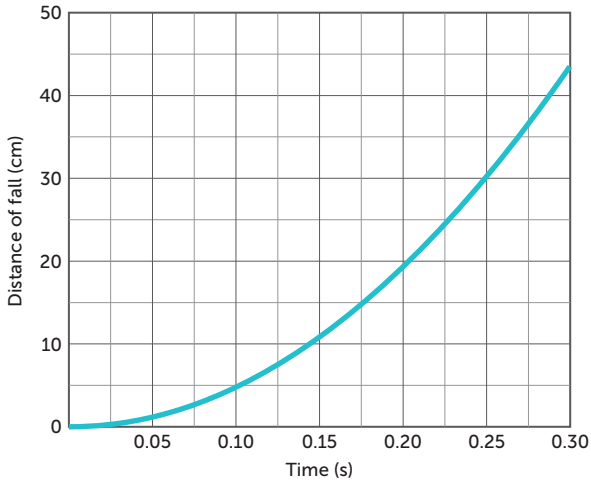
ACTIVITY 3.6 Investigating reaction time

Responses do not occur instantaneously; even reflex responses require time for perception of the stimulus, for conduction of impulses to and from the brain or spinal cord, and for the effector to carry out the response. Reaction time is the time it takes to respond to a stimulus. It depends on many factors, including the type and intensity of the stimulus and whether conscious thought is involved in the response.

Design and carry out an investigation into reaction time. Some things to consider are:

- What variable will you test? For example, you might test left vs right hand, different ages, different times of the day, males vs females, the effect of drinking caffeine, the effect of practice or of distraction.
- What is your hypothesis?
- What reactions will you test? For example, you might test how quickly a person is able to catch a falling ruler or to react to a light coming on.
- How will you measure the reaction time? You may be able to use an electronic device, a test on the Internet, or measure the distance of a falling object. (See the following graph for converting the distance into time.)
- How many trials will you conduct?
- What format will you use to record your results?
- How can you maximise the accuracy, reliability and validity of the investigation?
- What conclusion can you reach? Does it support your hypothesis?



Graph showing time taken for an object to fall a given vertical distance

ACTIVITY 3.7 Testing more reaction times

Reaction time is the time that elapses between a stimulus and the response to the stimulus. You can test your reaction time by following the weblink.

1. Describe the pathway taken by the nerve impulses involved in detecting the stimulus and making the response.
2. Is the response an innate or an acquired response?
3. Draw a column graph showing your reaction time for five trials.
4. Does your reaction time decrease with practice? If so, suggest why.
5. Do five trials with your left hand and then five trials with your right hand. Describe and explain any difference between the two sets of trials.

Reaction times

CHAPTER 3 SUMMARY

- Neurons are specialised cells that allow rapid communication within the body.
- Neurons are made up of:
  - a cell body, which contains organelles and controls the functioning of the cell
  - dendrites, which take messages to the cell body
  - axons, which take messages away from the cell body.
- Axons are surrounded by a myelin sheath that is produced by Schwann cells.
- The gaps between the myelin sheath are called nodes of Ranvier.
- The gap between adjacent neurons is called a synapse. Neurotransmitters cross the gap and transmit messages from one neuron to the next.
- Neurons can be classified as sensory, motor or interneurons based on their function.
- Neurons can also be classified as multipolar, bipolar, unipolar or pseudounipolar based on their structure.
- Axons are also known as nerve fibres. These are arranged in bundles that are surrounded by connective tissues. Multiple bundles are joined together to form a nerve.
- Messages travel along nerve fibres by an electrochemical change known as a nerve impulse.
- A potential difference is created by a separation of positive and negative charges. In the case of cells, it is due to ions creating a negative charge inside the cell and a positive charge outside the cell.
- The fluid outside the cell has a high concentration of positive sodium ions. The fluid inside the cell has a high concentration of positive potassium ions in addition to negative organic ions. This difference creates a resting membrane potential of –70 mV, meaning that the inside is 70 mV more negative than the outside.
- Ions are able to cross the membrane through leakage channels that are always open, or through gated channels that only open in response to a particular stimulus.
- Sodium and potassium ions are also moved across the membrane through a sodium–potassium pump, which moves three sodium ions out of the cell for every two potassium ions that move into the cell. This goes against the concentration gradient and, therefore, is active transport.
- An action potential is the rapid depolarisation and repolarisation of the cell membrane.
- Depolarisation is started by a stimulus from another neuron or a receptor. This stimulus opens some sodium ion gated channels, resulting in an influx of sodium ions that makes the inside of the cell more positive. If the membrane potential increases by 15 mV, then it reaches the threshold which leads to the opening of voltage-gated sodium ions, resulting in a greater flow of sodium ions into the cell. The inside of the cell becomes more positive than the outside, reaching approximately +40 mV.
- Repolarisation occurs due to the closing of the sodium channels and opening of potassium channels. The flow of sodium ions into the cell decreases and the flow of potassium ions out of the cell increases. This process makes the inside of the cell more negative than the outside, reducing the membrane potential.
- An action potential in one area of the membrane stimulates an action potential in the adjacent area. Thus, the action potential moves along the nerve fibre in a nerve impulse.
- The sodium ion voltage-gated channels become inactivated after they open, and so they cannot be stimulated for the period during and shortly after the action potential, the refractory period. This means that a nerve impulse cannot travel backwards.

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- Myelin acts as an insulator on the cell membrane, preventing the formation of an action potential. Therefore, the action potentials jump from one node of Ranvier to the next. This is called saltatory conduction and means that the nerve impulse travels much faster in myelinated fibres than in unmyelinated fibres.
- Nerve impulses are always the same strength. They can, however, differ in the frequency of impulses and the number of nerve fibres affected. This allows us to differentiate between stimuli.
- When a nerve impulse reaches a synapse, calcium ion voltage-gated channels are opened, allowing calcium ions to flow into the cell. This stimulates the release of neurotransmitters by exocytosis. The neurotransmitters diffuse across the synapse and bind to receptors on the next neuron, stimulating an action potential.
- There are different receptors to detect different stimuli, including thermoreceptors, osmoreceptors, chemoreceptors, pain receptors and touch receptors.
- Reflexes are a rapid, automatic response to a change in environment. They often protect the body from harm, and may be innate or learnt.
- Spinal reflexes are carried out without input from the brain. They involve a stimulus, a receptor, a sensory neuron, possibly one or more interneurons, a motor neuron and an effector.
- The endocrine and nervous systems both transmit messages through the body.
- The nervous system reacts quickly and affects specific targets, but the effect is short lasting.
- The endocrine system is slower to react and generally has a broader target, but the effect is longer lasting.

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CHAPTER 3 GLOSSARY

Acquired reflex A response to a stimulus that has been learnt through practice

Action potential The rapid depolarisation and repolarisation of the cell membrane

Afferent neuron *see* sensory neuron

All-or-none response A response of a constant size regardless of the strength of the stimulus; with respect to nerve cells, a nerve impulse is transmitted at full strength or not at all

Association neuron *see* interneuron

Axon An extension of the cell body of a nerve cell; carries nerve impulses away from the cell body

Axon terminal The end of a branch of the axon

Bipolar neuron A neuron with two processes – one axon and one dendrite – arising from opposite sides of the cell body; these neurons are sensory, such as the neurons found in the retina of the eye

Cell body The part of a neuron that contains the nucleus

Chemoreceptor A receptor sensitive to particular chemicals

Connector neuron *see* interneuron

Core temperature The temperature inside the body

Dendrite An extension of the body of a nerve cell; carries nerve impulses into the cell body

Depolarisation The process of becoming depolarised

Depolarised Describes the membrane of a nerve cell when there is no difference in electrical charge between the inside and outside of the membrane

Effector neuron *see* motor neuron

Efferent neuron *see* motor neuron

Electrochemical change The change in electrical voltage brought about by changes in the concentration of ions inside and outside the cell membrane of a neuron

Extracellular fluid Fluid outside the body cells; includes tissue fluid and blood plasma

Hyperpolarisation The process of becoming hyperpolarised

Hyperpolarised Describes the membrane of a nerve cell when it has a lower membrane potential than normal

Innate reflex A response to a stimulus that is acquired genetically and is therefore present at birth

Interneuron A nerve cell in the brain or spinal cord that carries messages between other nerve cells; also called association neuron, connector neuron or relay neuron

Intracellular fluid The fluid inside cells

Ion A charged atom or molecule

Leakage channel A protein channel that is always open

Membrane potential The electrical voltage across the membrane of a cell (usually a nerve cell)

Motor neuron A nerve cell that carries messages from the brain or spinal cord to effector organs (muscles and glands); also called an effector neuron

Multipolar neuron A nerve cell with one axon and many dendrites; the most common type of neuron

Myelin sheath A white, fatty sheath that surrounds some nerve fibres

Myelinated fibre A nerve fibre that has a myelin sheath

Nerve A bundle of nerve fibres

Nerve fibre A projection from a nerve cell body with its associated coverings; usually refers to an axon

Nerve impulse The electrochemical change that travels along the membrane of a nerve cell; the message carried by a nerve

Neurilemma A sheath surrounding a nerve fibre

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Neuromuscular junction The junction between branches of a motor nerve cell and a muscle fibre; also called the motor end plate

Neuron A nerve cell; the basic structural and functional unit of the nervous system

Neurotransmitter A molecule that carries a nerve impulse across the small gap between branches of adjacent nerve cells

Node of Ranvier A gap in the myelin sheath of a nerve fibre

Osmoreceptor A receptor sensitive to osmotic pressure of body fluids

Pain receptor A receptor that is stimulated by damage to tissues

Polarised Describes the situation when the inside of the membrane of a nerve cell has a negative electrical charge compared with the outside

Potential difference A difference in electrical charge between two locations

Pseudounipolar neuron A neuron with a single process that splits into two; the cell body is to one side of the axon; in humans, such neurons are sensory and carry messages to the spinal cord

Receptor A structure that detects a stimulus

Receptor neuron *see* sensory neuron

Reflex A rapid, automatic response to a change in the external or internal environment; tries to restore homeostasis

Reflex arc The pathway travelled by nerve impulses from receptor to effector in a reflex

Refractory period A short period following a stimulus during which a nerve cell or a muscle fibre cannot be stimulated again

Relay neuron *see* interneuron

Repolarisation The process of becoming repolarised

Repolarised A membrane that has returned to a polarised state

Resting membrane potential The membrane potential of unstimulated nerve cells

Saltatory conduction The conduction of a nerve impulse along a myelinated nerve fibre; the impulse seems to jump from one node of Ranvier to the next

Schwann cell A cell that wraps around a nerve fibre, forming the myelin sheath

Sense organ Receptors grouped into a discrete organ such as the eye

Sensory neuron A nerve cell that carries messages from receptors to the brain or spinal cord; also called a receptor neuron

Sodium–potassium pump A mechanism in the membrane of a nerve cell that transports sodium ions out of the cell and potassium ions into the cell by active transport

Spinal reflex A reflex carried out by the spinal cord without involvement of the brain

Spinal reflex arc The pathway travelled by a nerve impulse from receptor to effector in a spinal reflex

Stimulus Any change, internal or external, that causes a response (plural: stimuli)

Synapse The junction between the branches of adjacent neurons

Thermoreceptor A temperature receptor; located in the skin or the hypothalamus

Threshold The potential after which an all-or-none response occurs

Touch receptor A receptor sensitive to touch

Unipolar neuron A neuron with a single process, an axon; does not occur in humans

Unmyelinated fibre A nerve fibre that has no myelin sheath

Voltage-gated channel A protein channel that is opened by an electrical stimulus

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CHAPTER 3 REVIEW QUESTIONS

Recall

1 Describe how the sheath of a myelinated fibre is formed.

2 One way that neurons can be classified is based on their function. Name the three types of neurons and describe the function of each.

3 a Define 'electrical potential'.
  b What is the potential of the membrane of a nerve cell when it is not conducting a nerve impulse?

4 a Define 'action potential'.
  b Formation of an action potential is an all-or-none response. Define 'all-or-none response'.

5 What is the 'refractory period' of an action potential?

6 a Explain how a nerve impulse passes along a nerve fibre.
  b Explain the difference between the way a nerve impulse is conducted along a myelinated and an unmyelinated nerve fibre.

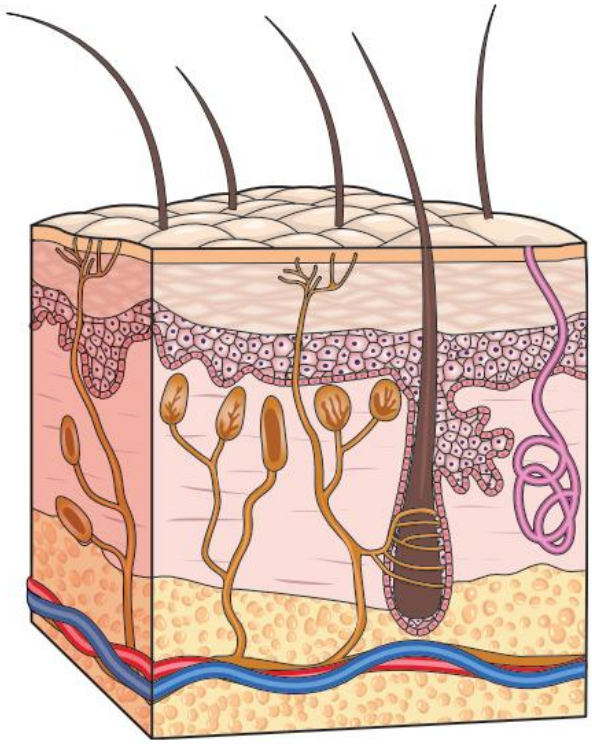
7 a What is a synapse?
  b Describe how a nerve message is carried across a synapse.

8 What is the difference between a synapse and a neuromuscular junction?

9 a Describe three differences between the action of nerves and that of hormones.
  b Describe some of the similarities between the nervous and endocrine systems.

10 In what parts of the body are thermoreceptors found?

11 In the diagram below, identify the receptors that would be stimulated by:
  a light touch
  b pressure or vibration
  c movement of hairs
  d pain.



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Explain

12 Explain the difference between a myelinated fibre and an unmyelinated fibre.

13 a Explain the difference between multipolar, bipolar, unipolar and pseudounipolar neurons.
   b Give an example of where each of these can be found.

14 Explain the difference between a neuron, a nerve and a nerve fibre.

15 Explain how the potential of a resting nerve cell membrane is maintained.

16 Explain the role of calcium ions in the transmission of a nerve impulse across a synapse.

17 Explain why a nerve impulse can only cross a synapse in one direction.

18 Explain how we are able to distinguish between a light touch and heavy pressure on the skin.

Apply

19 In what ways do nerve cells differ from most body cells?

20 A nerve impulse is often described as an electrochemical change. Explain why it is described in this way.

21 Hyperkalaemia is a higher-than-normal level of potassium in the blood and therefore in the extracellular fluid. What effect would hyperkalaemia have on the resting membrane potential of nerve cells?

22 In an examination a student stated that 'an action potential is another name for a nerve impulse'. Is this statement correct? Explain your answer.

23 Lightly press a pencil point on to the skin of your palm. Gradually increase the force with which you are pushing the pencil. How are you able to distinguish different intensities of the same stimulus?

24 The speed of transmission of nerve impulses can vary from 2 m/s to 140 m/s. Explain how there can be such a wide range of speeds of transmission of impulses.

25 Name the type of receptor that would recognise:
   a an increase in carbon dioxide in the blood
   b a graze on an elbow
   c a light breeze blowing.

26 Many reflexes are protective. List five protective reflexes.

27 A driver approaching traffic lights saw the lights change from green to amber. She transferred her foot from the accelerator to the brake in order to stop. Describe the pathways of the nerve impulses that would be involved in this response.

28 When you withdraw your hand from a painful stimulus, the response occurs before you become consciously aware that you have hurt yourself. Explain how this is possible.

29 Why would it be unwise to continually take painkillers for a particular pain without seeking medical help?

Extend

30 Multiple sclerosis is caused by destruction of the myelin sheath. Use references to find out how damage to the sheath results in the jerky body and limb movements, double vision, slurred speech and paralysis that may occur as a result of the disease.

31 Doctors may use reflexes to find out whether a patient has an impairment of the nervous system. Absence or exaggeration of a particular reflex may indicate damage to nerves or to the spinal cord through injury or disease. Conduct research to find out about the following reflexes and what absence or exaggeration of the reflex could indicate:
   a patellar reflex (knee jerk)
   b Achilles reflex (ankle jerk)
   c abdominal reflex
   d plantar reflex and Babinski sign.

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4 THE NERVOUS SYSTEM IS HIGHLY ORGANISED

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

» select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions

SCIENCE UNDERSTANDING

Central and peripheral nervous system

» structure and function of the divisions of the nervous system can be observed and compared at different levels in detecting and responding to the changes in the internal and external environments, including:
  – central–peripheral
  – afferent–efferent
  – autonomic–somatic
  – sympathetic–parasympathetic

» the parts of the central nervous system, including the brain (cerebrum, cerebellum, medulla oblongata, hypothalamus, corpus callosum) and spinal cord, have specific roles in the co-ordination of body functions and are protected by the meninges and cerebro-spinal fluid

Source: School Curriculum and Standards Authority, Government of Western Australia

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The trillions of cells that make up the human body work together in an integrated and coordinated way. To achieve integration and coordination, cells must communicate with one another. The **nervous system** is the communication network and control centre of the body and is made up of the brain, spinal cord and nerves. While it can be divided into parts, or divisions, on the basis of their structure and functions – the **central nervous system (CNS)** consists of the brain and spinal cord; and the **peripheral nervous system (PNS)** is made up of the nerves that connect the CNS with the receptors, muscles and glands – these all work together in a coordinated way.

4.1 CENTRAL NERVOUS SYSTEM

The central nervous system is where incoming messages are processed and outgoing messages are initiated.

Protection of the central nervous system

The brain and the spinal cord are very delicate and vital parts of the body. Therefore, it is important that they are well protected. Three structures protect the CNS:

- bone
- membranes called meninges
- cerebrospinal fluid.

Cranium and vertebrae

The outermost protective layer is bone. The brain is protected by the **cranium**, the part of the skull that houses the brain, while the spinal cord runs through the **vertebral canal**, an opening in the vertebrae. These bones provide a strong, rigid structure to protect the structures underneath.

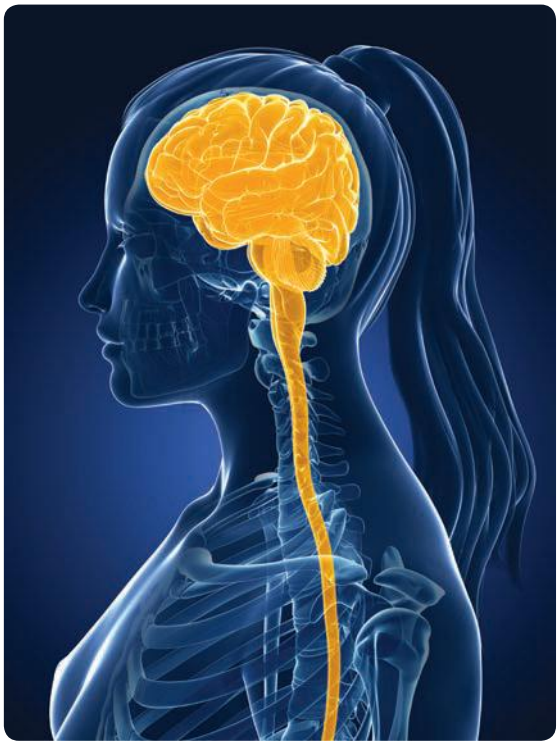

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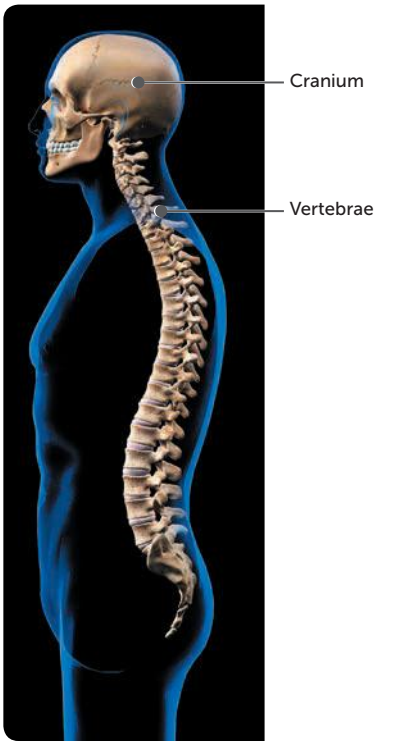
FIGURE 4.1 The central nervous system – brain and spinal cord



Alamy Stock Photo/Stocktrek Images, Inc.

FIGURE 4.2 The cranium and vertebrae protect the brain and spinal cord

Meninges

Inside the bones, and covering the surface of the brain and spinal cord (i.e. the entire CNS), are three layers of connective tissue forming membranes called the **meninges**.

- The outer meningeal layer, the **dura mater**, is tough and fibrous, and therefore provides a layer of protection for the brain. It sticks closely to the bones of the skull, but on the inside of the vertebral canal it is not so close fitting. This outer membrane has been described as having a texture and thickness similar to a household rubber glove.
- The middle meningeal layer, the **arachnoid mater**, is a loose mesh of fibres.
- The inner layer, the **pia mater**, is far more delicate. It contains many blood vessels and sticks closely to the surface of the brain and spinal cord.

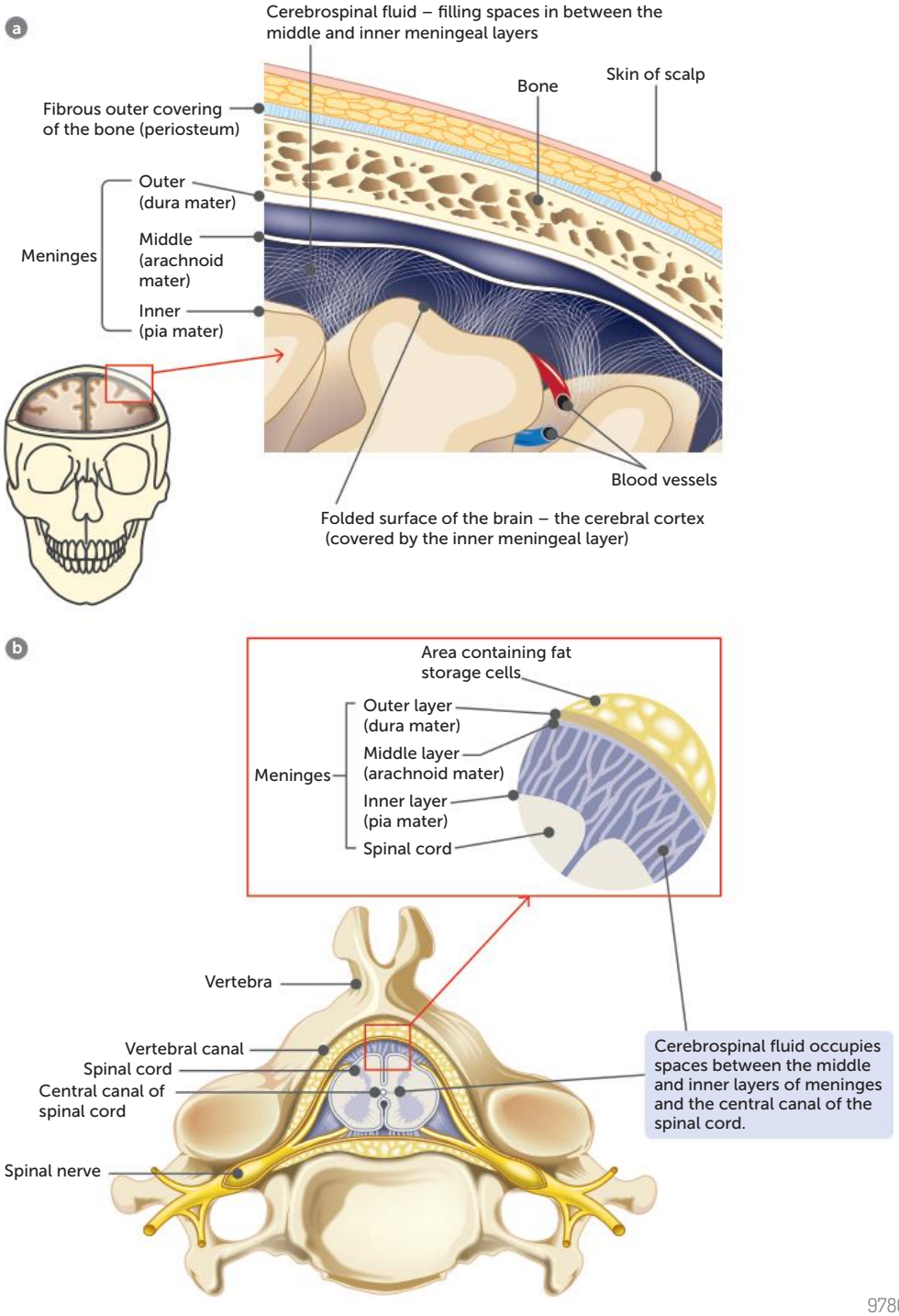


FIGURE 4.3
a Structures that protect the brain; **b** Structures that protect the spinal cord

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Cerebrospinal fluid

The third protective structure is **cerebrospinal fluid (CSF)**, which occupies a space between the middle and inner layers of meninges. It also circulates through cavities in the brain and through a canal in the centre of the spinal cord. The CSF is a clear, watery fluid containing a few cells and some glucose, protein, urea and salts.

The CSF has three functions:

- *Protection:* the CSF acts as a shock absorber, cushioning any blows or shocks the CNS may sustain.
- *Support:* the brain is suspended inside the cranium and floats in the fluid that surrounds it.
- *Transport:* the CSF is formed from the blood, and circulates around and through the CNS before eventually re-entering the blood capillaries. During its circulation it takes nutrients to the cells of the brain and spinal cord and carries away their wastes.

Key concept

The central nervous system is protected by bones, meninges and cerebrospinal fluid.

The brain

The brain is a very complex organ, both in structure and function. Much of the brain's workings are still a mystery and new discoveries are constantly being made. In this section we will deal only with those parts of the brain that have major functions, but it is important to remember that the brain works as an integrated whole.

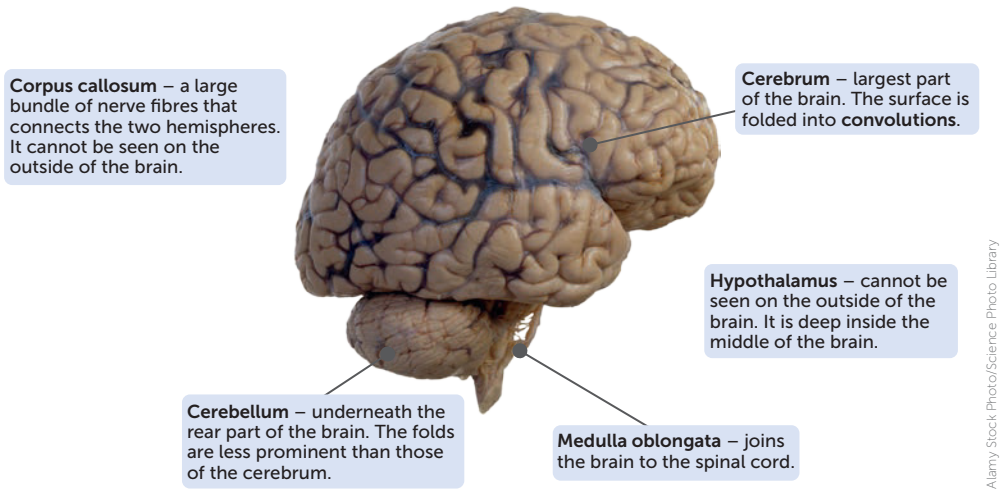
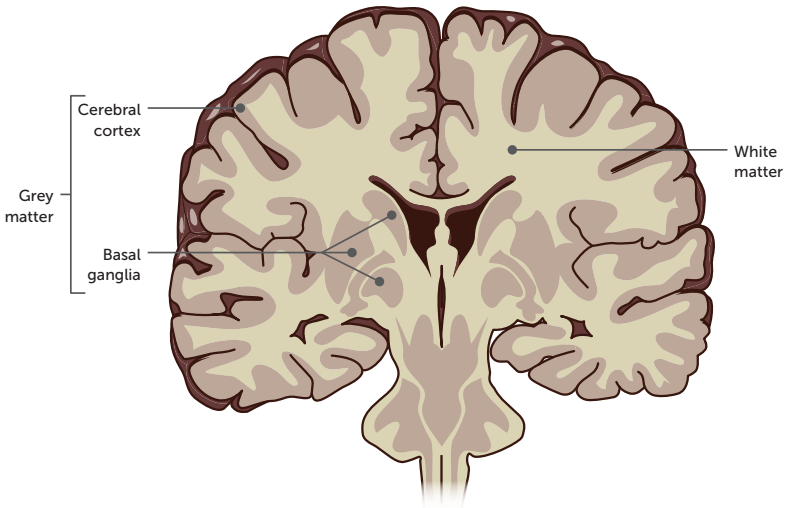


FIGURE 4.4 External view of the brain

Cerebrum

The **cerebrum** is by far the biggest part of the brain. It consists of an outer surface about 2–4 mm thick of **grey matter** known as the **cerebral cortex**. The grey matter consists of neuron cell bodies, dendrites and unmyelinated axons. Below the cortex is **white matter**, which is made up of myelinated axons. The fatty nature of myelin gives the white matter its colour and texture. Deep inside the cerebrum is additional grey matter called the **basal ganglia**.

FIGURE 4.5 Cross-section of the cerebrum



The cerebral cortex is folded in patterns that greatly increase its surface area. In this way the cortex contains 70% of all the neurons in the central nervous system. The folding produces rounded ridges called **convolutions** (or gyri; singular: **gyrus**). The convolutions are separated by either shallow downfolds called **sulci** (singular: sulcus) or deep downfolds called **fissures**.

FIGURE 4.6 Convolutions, sulci and fissures of the cerebrum, as seen from above

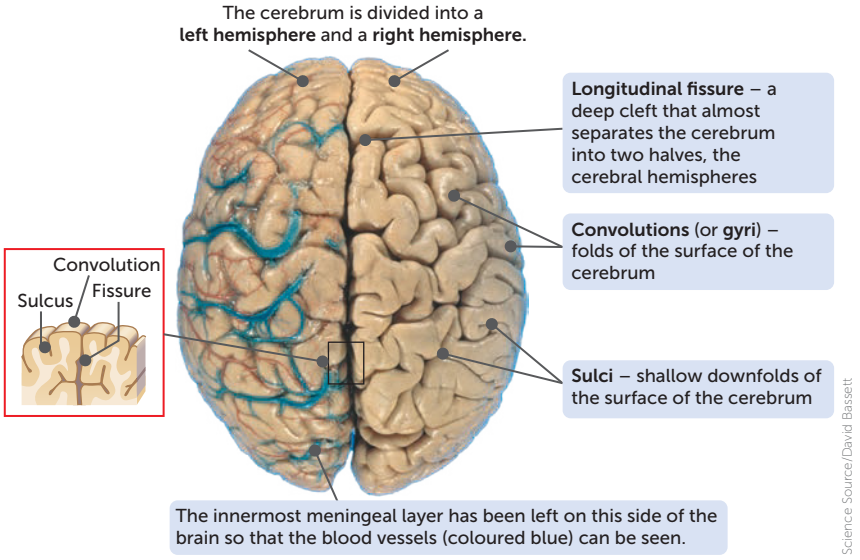


TABLE 4.1 Functions of the lobes of the cerebral cortex

LOBE	FUNCTION
Frontal lobe	Thinking, problem solving, emotions, personality, language, and control of movement
Parietal lobe	Processing temperature, touch, taste, pain and movement
Temporal lobe	Processing memories and linking them with senses; receives auditory information
Occipital lobe	Vision
Insula	Recognition of different senses and emotions, addiction and psychiatric disorders

The deepest fissure, the **longitudinal fissure**, almost separates the cerebrum into two halves – the left and right **cerebral hemispheres**. Joining the two hemispheres, at the base of the longitudinal fissure, is an area of white matter consisting of a large bundle of transverse fibres known as the corpus callosum.

The patterns of folding of the cerebral cortex vary from person to person. However, certain fissures and sulci are fairly constant and are used to further subdivide each cerebral hemisphere into four lobes – the **frontal**, **temporal**, **occipital** and **parietal lobes**. Another part of the cerebrum, the **insula**, is deep inside the brain and is regarded as a fifth lobe.

The cerebral cortex is involved in mental activities such as thinking, reasoning, learning,

memory, intelligence and sense of responsibility. It is also concerned with perception of the senses and the initiation and control of voluntary muscle contraction. Nearly all the impulses from our sense organs are carried to the cerebral cortex, which then has all the relevant information about the environment and can initiate responses accordingly.

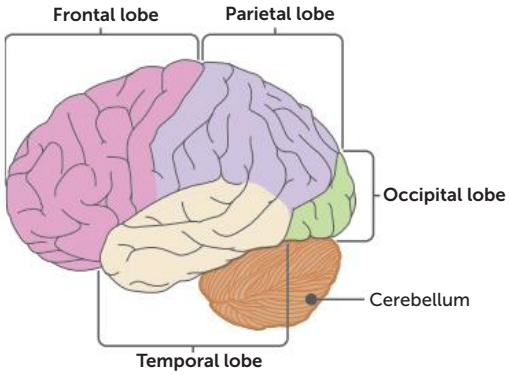


FIGURE 4.7 The lobes of the cerebrum. The fifth lobe, the insula, is not visible from the outside of the brain

The cortex can be roughly divided into three functional areas.

- **sensory areas**, which interpret impulses from receptors
- **motor areas**, which control muscular movements
- **association areas**, which are concerned with intellectual and emotional processes.

One of the important functions of the cerebrum is memory. The association areas of the cerebral cortex are involved in memory. Memories are not stored in individual memory cells in the brain: they are pathways of nerve cells. When a memory is stored, new links are made between neurons or existing links are modified.

Although the two sides of the cerebrum appear to be very similar, close inspection shows that they are not identical. For example, in right-handed people the right frontal lobe is wider than the left and the left occipital and parietal lobes are wider than the right ones. Many specialised functions occur in only one hemisphere. Language ability, for example, is normally controlled by the left hemisphere; musical and artistic abilities are functions of the right hemisphere.

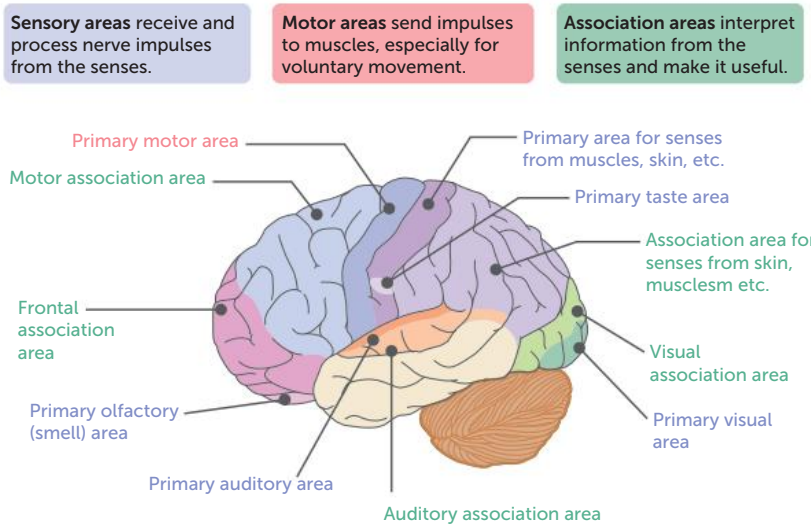


FIGURE 4.8 Some functional areas of the cerebral cortex

Cerebral hemispheres
This website provides further information on the different functions of the two cerebral hemispheres.

Between the cerebral cortex and the basal ganglia is white matter composed of bundles of nerve fibres that are surrounded by a sheath of white fatty material called **myelin**. Within the CNS, bundles of nerve fibres are called **tracts**; outside the CNS they are called nerves.

Three types of tracts occur in the white matter:

- tracts that connect various areas of the cortex within the same hemisphere
- tracts that carry impulses between the left and right hemispheres
- tracts that connect the cortex to other parts of the brain or to the spinal cord.

The basal ganglia consist of groups of nerve cell bodies associated with control of skeletal muscles. They play a role in initiating desired movements and inhibiting unwanted movement.

Key concept

The cerebrum is responsible for thinking, reasoning, learning, memory and sense of responsibility. Its folded structure increases the surface area for a large number of neurons.

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Corpus callosum

The **corpus callosum** is a wide band of nerve fibres that lies underneath the cerebrum at the base of the longitudinal fissure. Nerve fibres in the corpus callosum cross from one cerebral hemisphere to the other and allow the two sides of the cerebrum to communicate with each other.

Cerebellum

The **cerebellum** lies under the rear part of the cerebrum. It is the second-largest part of the brain and its surface is folded into a series of parallel ridges. The outer folded part of the cerebellum is grey matter. Inside is white matter that branches to all parts of the cerebellum, rather like the branches of a tree.

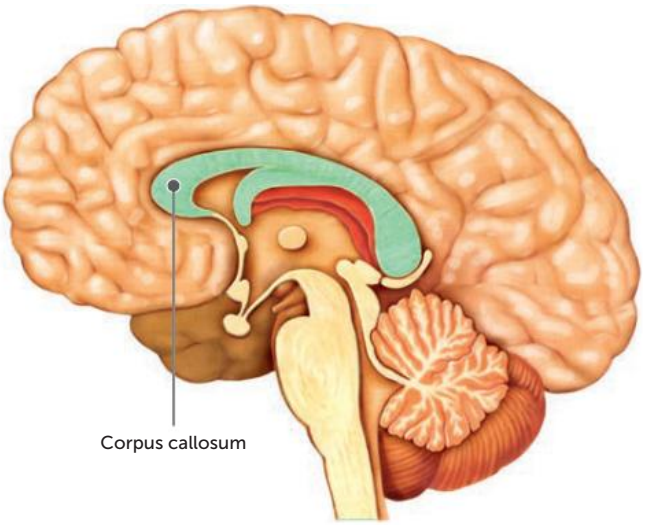

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FIGURE 4.9 The corpus callosum joins the left and right hemispheres of the cerebrum

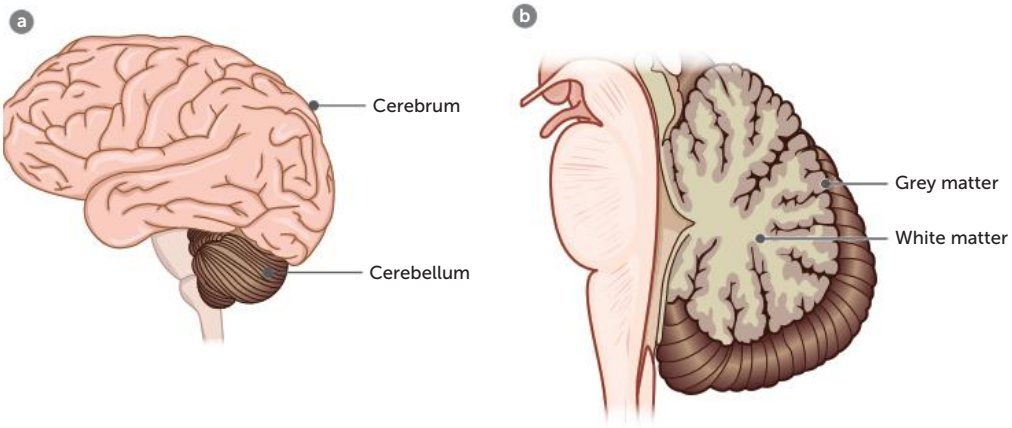

FIGURE 4.10
a Location of the cerebellum;
b Cross-section of the cerebellum

The cerebellum exercises control over posture, balance and the fine coordination of voluntary muscle movement. To carry out these functions the cerebellum receives sensory information from:

- the inner ear for information about posture and balance
- stretch receptors in the skeletal muscles for information about the length of muscles.

All the functions of the cerebellum take place below the conscious level. Impulses do not originate in the cerebellum and so without it we could still move, but our movements would be spasmodic, jerky and uncontrolled. Smooth, coordinated movements, such as those required for writing, playing a musical instrument or using a computer, would be impossible.

Key concept

The cerebellum lies at the back of the brain and is responsible for posture, balance and the coordination of movement.

Hypothalamus

The **hypothalamus** lies in the middle of the brain and cannot be seen from the outside. Although small, the hypothalamus controls many bodily activities, but it is mostly concerned with maintaining a constant internal environment – homeostasis.

The functions of the hypothalamus include the regulation of:

- the autonomic nervous system, including the regulation of heart rate, blood pressure, the secretion of digestive juices, movements of the alimentary canal and the diameter of the pupil of the eye
- body temperature
- food and water intake
- patterns of waking and sleeping
- contraction of the urinary bladder
- emotional responses, such as fear, anger, aggression, pleasure and contentment
- the secretion of hormones and coordination of parts of the endocrine system; acting through the pituitary gland, the hypothalamus regulates metabolism, growth, reproduction and responses to stress.

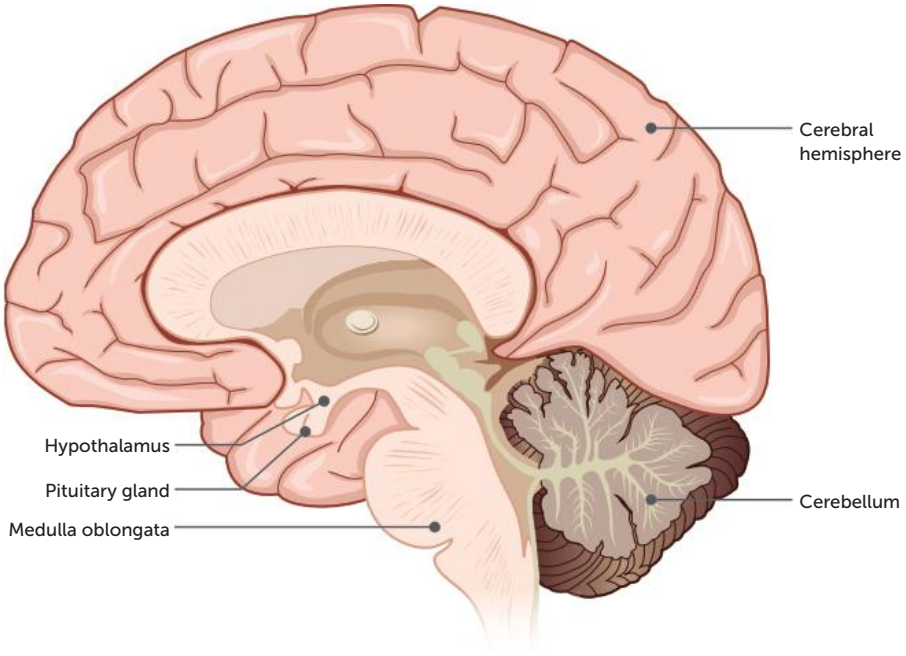

FIGURE 4.11 Location of the hypothalamus and medulla oblongata

Medulla oblongata

The **medulla oblongata** is a continuation of the spinal cord. It is about 3 cm long and extends from just above the point where the spinal cord enters the skull (see Figure 4.11). Many nerve fibres simply pass through the medulla going to or from the other parts of the brain, but the medulla does have an important role in automatically adjusting body functions.

The medulla oblongata contains:

- the **cardiac centre**, which regulates the rate and force of the heartbeat
- **respiratory centres**, which control rate and depth of breathing
- the **vasomotor centre**, which regulates the diameter of blood vessels.

In addition, other centres regulate the reflexes of swallowing, sneezing, coughing and vomiting. All the centres in the medulla oblongata are influenced and controlled by higher centres in the brain, particularly the hypothalamus.

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Spinal cord

The **spinal cord** is a roughly cylindrical structure about 44 cm long in an adult. It extends from the foramen magnum, the large opening at the base of the skull, to the second lumbar vertebra, which is at about waist level.

As we have seen, the spinal cord, like the brain, is heavily protected. The cord is enclosed in the vertebral canal, and inside the ring of bone are the three meningeal layers. However, the outermost meningeal layer is not joined to the bone as it is in the skull. Instead, a space containing fat, connective tissue and blood vessels serves as padding around the spinal cord and allows the cord to bend when the spine is bent.

If a cross-section of the spinal cord is examined, it is seen to consist of areas of grey matter and areas of white matter. The composition of these two areas is the same as in the brain – the grey matter is composed of nerve cell bodies and unmyelinated nerve fibres, and the white matter is composed of myelinated fibres. Unlike the cerebrum and cerebellum of the brain, where the grey matter is at the surface, the grey matter of the spinal cord is at the centre, surrounded by the white matter.

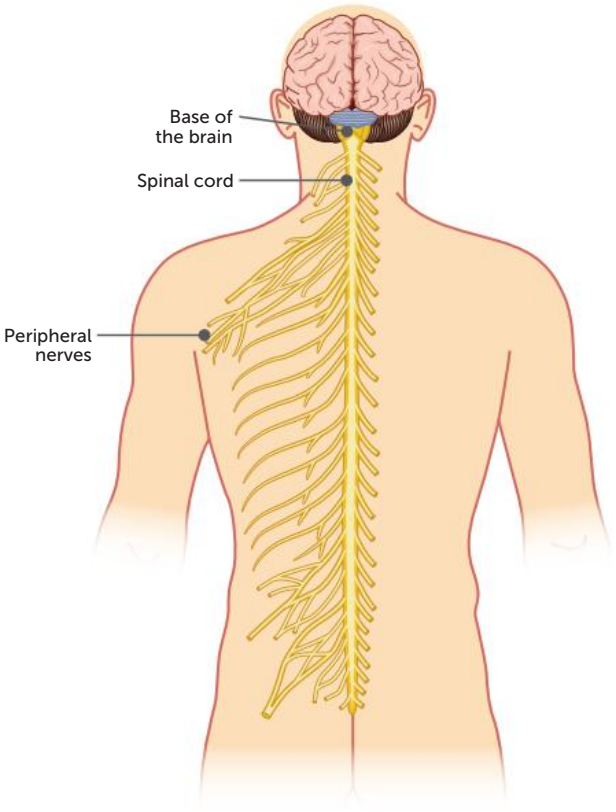

FIGURE 4.12 Position of the spinal cord

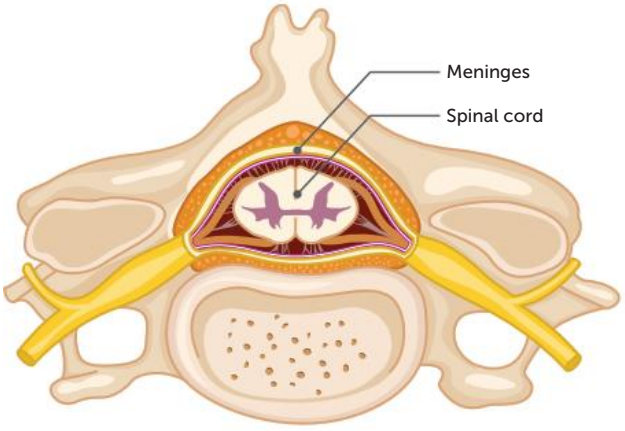

FIGURE 4.13 Cross-section of a vertebra showing the position of the spinal cord and meninges

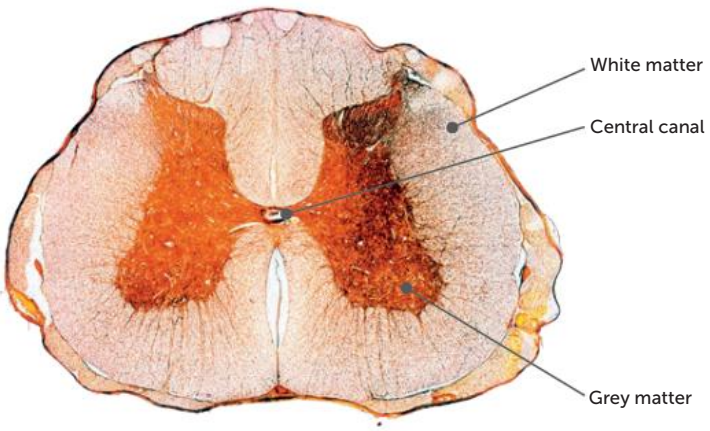

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FIGURE 4.14 Cross-section of the spinal cord as seen under the low power of a microscope. The grey matter is coloured brown in this photo

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The grey matter is roughly in the shape of a letter H. In the cross-bar of the H is a small space called the **central canal**, which runs the length of the spinal cord and contains cerebrospinal fluid.

The myelinated nerve fibres of the white matter are arranged in bundles known as ascending and descending tracts.

- **Ascending tracts** are sensory axons that carry impulses upwards, towards the brain.
- **Descending tracts** contain motor axons that conduct impulses downwards, away from the brain.

Thus, one of the functions of the spinal cord is to carry sensory impulses up to the brain and motor impulses down from the brain. The second function of the spinal cord is to integrate certain fast, automatic responses called reflexes. The mechanism involved in spinal reflexes was discussed in Chapter 3.

Key concept

The spinal cord takes messages between the brain and the peripheral nervous system.

TABLE 4.2 Structure and functions of the main areas of the CNS

STRUCTURE	FUNCTION
Cerebral cortex	Higher-order functions such as thinking, reasoning, memory, learning, conscious awareness of surroundings
Corpus callosum	Communication between the two cerebral hemispheres
Cerebellum	Coordination of fine contractions of muscles resulting in smooth movements and the maintenance of posture and balance
Hypothalamus	Homeostasis; regulation of the heart, digestive system, appetite, thirst, metabolism, body temperature, response to fear or anger
Medulla oblongata	Under the influence of the hypothalamus, regulates the heart, breathing and diameter of blood vessels
Spinal cord	Provides a pathway for communication between muscles and glands and the brain; integration of automatic, protective reflexes

Spinal cord
This website provides more information about the spinal cord.

Activity 4.1
Examining a dissected brain

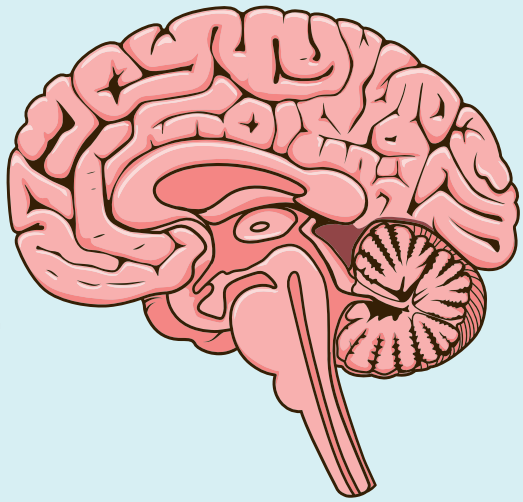
Activity 4.2
Phineas Gage

Questions 4.1

RECALL KNOWLEDGE

1 Label the following structures on the diagram below.

a Cerebellum
b Hypothalamus
c Pituitary gland
d Cerebrum
e Medulla oblongata
f Corpus callosum
g Spinal cord
h Convolution
i Sulci



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2 Explain how the central nervous system is different from the peripheral nervous system.
3 List the structures that protect the central nervous system.
4 Explain how cerebrospinal fluid protects the brain.
5 a Label the parietal, frontal, occipital and temporal lobes of the cerebral cortex on the diagram below.

b Explain why it is not possible to label the insula in this diagram.
6 Describe the functional areas of the cerebrum.
7 Describe the appearance of a cross-section of the cerebellum.
8 Name and describe the centres that are located in the medulla oblongata.
9 Compare and contrast the structure of the brain and spinal cord.
10 Draw a labelled cross-section of the spinal cord to show the grey matter, white matter and central canal.

APPLY KNOWLEDGE

11 Explain why a broken back can result in the inability to move limbs.
12 Justify the naming of the grey matter and white matter.
13 Describe the movement of someone without a cerebellum. Justify your prediction using your knowledge of the functions of the cerebellum.
14 Suggest a reason for the hypothalamus being located towards the centre of the brain.

WS
4.1 Central nervous system

4.2 PERIPHERAL NERVOUS SYSTEM

The peripheral nervous system takes messages from receptors to the central nervous system and from the CNS to muscles and glands. It is composed of:

- nerve fibres that carry information to and from the CNS
- groups of nerve cell bodies, called **ganglia**, which lie outside the brain and spinal cord.

Types of nerves

The nerve fibres are arranged into nerves, which arise from the brain and the spinal cord.

Cranial nerves

Twelve pairs of nerves, such as the optic nerve and auditory nerve, arise from the brain. These are the **cranial nerves**. Most cranial nerves are mixed nerves; that is, they contain fibres that carry impulses into the brain, as well as fibres that carry impulses away from the brain. Fibres that carry impulses into the CNS are called **sensory fibres**; those that carry impulses away from the CNS are **motor fibres**. A few cranial nerves carry only sensory impulses or only motor impulses.

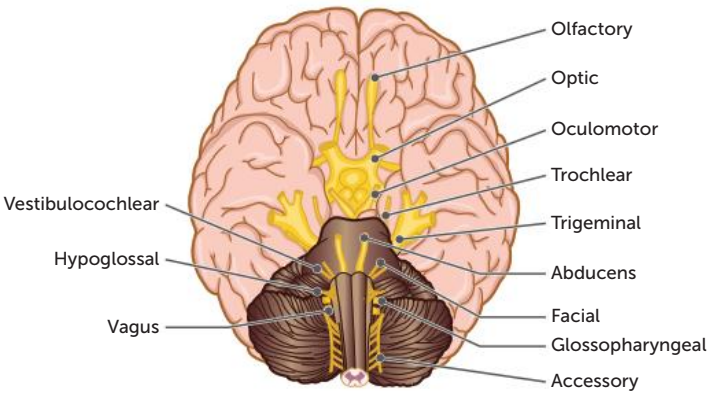

FIGURE 4.15 The 12 pairs of cranial nerves

Spinal nerves

Thirty-one pairs of **spinal nerves** arise from the spinal cord. They are all mixed nerves containing both sensory and motor fibres. Each nerve is joined to the spinal cord by two roots. The **ventral root** contains the axons of motor neurons that have their cell bodies in the grey matter of the spinal cord. The **dorsal root** contains the axons of sensory neurons that have their cell bodies in a small swelling on the dorsal root known as the **dorsal root ganglion**.

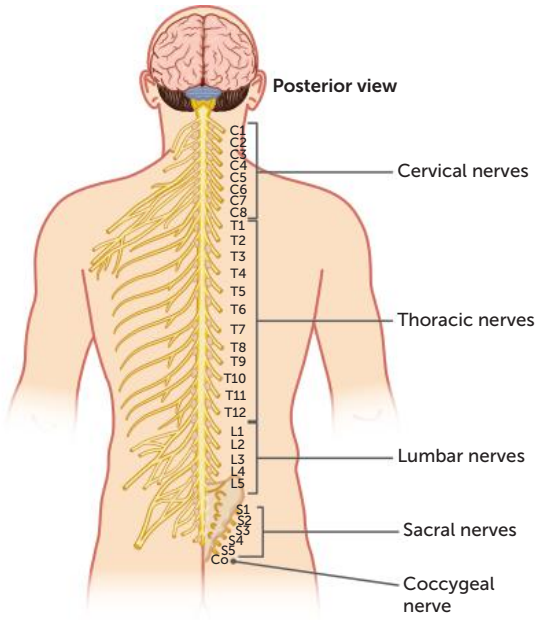

FIGURE 4.16 The 31 pairs of spinal nerves

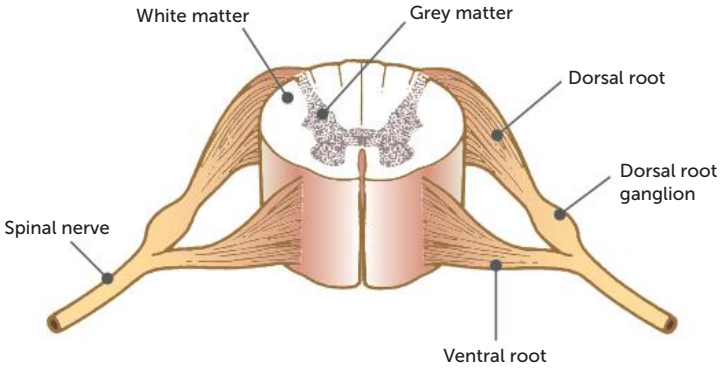

FIGURE 4.17 Cross-section of the spinal cord, showing a pair of spinal nerves with the dorsal and ventral roots

Key concept

The peripheral nervous system is made up of 12 pairs of cranial nerves and 31 pairs of spinal nerves that take messages between the central nervous system and receptors, muscles and glands.

Divisions of the peripheral nervous system

The nerves that make up the PNS contain fibres carrying nervous impulses to and from all parts of the body. To make it easier to study the various functions of the PNS, it has been divided and subdivided into parts, each with a particular function.

Afferent division

The **afferent** (or sensory) **division** of the PNS has fibres that carry impulses *into* the CNS by sensory neurons from receptors in the skin and around the muscles and joints. These neurons can be further divided into:

- **somatic sensory neurons**, which bring impulses from the skin and muscles
- **visceral sensory neurons**, which bring impulses from the internal organs.

Efferent division

The **efferent** (or motor) **division** has fibres that carry impulses *away* from the CNS. It is subdivided into:

- the **somatic division** (sometimes called the somatic nervous system), which takes impulses from the CNS to the skeletal muscles
- the **autonomic division** (autonomic nervous system), which carries impulses from the CNS to heart muscle, involuntary muscles and glands. The autonomic division is further subdivided into the:
  - **sympathetic division** (sympathetic nervous system)
  - **parasympathetic division** (parasympathetic nervous system).

These parts of the nervous system are summarised in Figure 4.18.

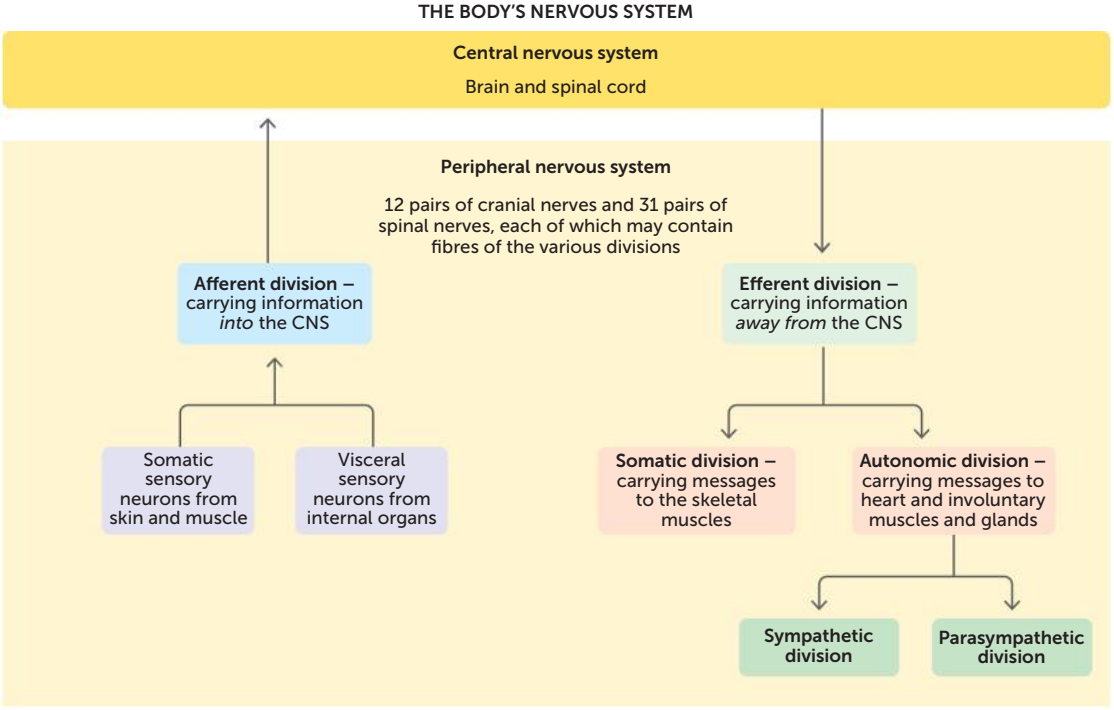

FIGURE 4.18 Functional organisation of the body's nervous system

Key concept

The peripheral nervous system is divided into divisions based on the type of nerve fibres, where the messages are travelling to or from, and the type of responses brought about.

Autonomic nervous system

The **autonomic nervous system (ANS)** controls the body's internal environment and is involved in many of the mechanisms that keep it constant. It usually operates without conscious control and is regulated by groups of nerve cells in the medulla oblongata, hypothalamus and cerebral cortex. Some of the body functions regulated by the autonomic division include:

- heart rate
- blood pressure
- body temperature
- digestion
- release of energy
- pupil diameter
- air flow to the lungs
- defecation
- urination.

The nerve fibres of the ANS make up part of the spinal nerves and part of some of the cranial nerves. They carry impulses to the heart muscle, other muscles of the internal organs and the glands. The impulse travels along two neurons from the CNS to an organ controlled by the ANS. The first neuron is myelinated and has its cell body in the CNS. The second neuron is unmyelinated and has its cell body in a ganglion (plural: ganglia); a group of nerve cell bodies outside the CNS.

The pathway from the CNS to heart muscle, involuntary muscle or glands is an important difference between the autonomic division and the somatic division. Where there are two motor neurons involved in the autonomic pathway, the somatic division has just one motor neuron carrying impulses from the CNS to the effector.

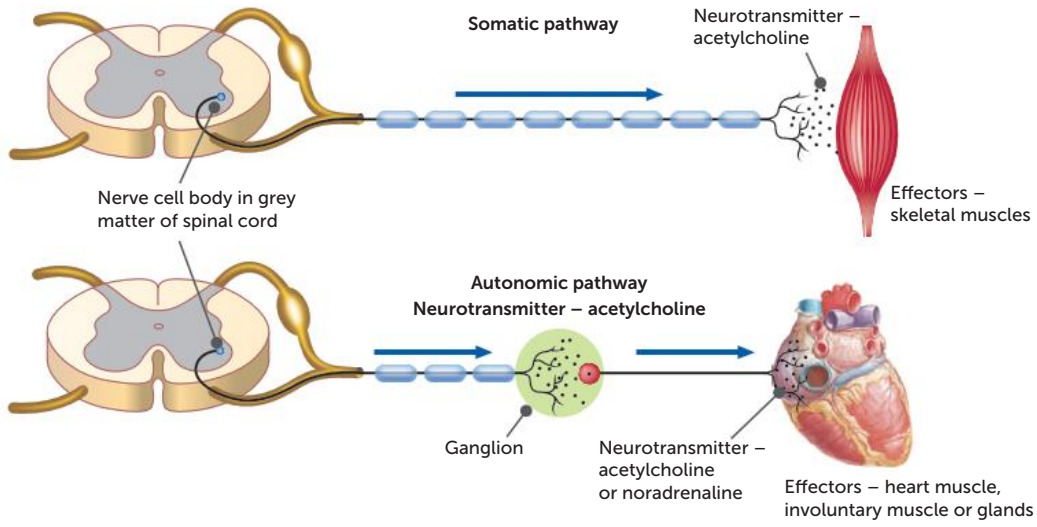

FIGURE 4.19 The difference in motor pathways between the autonomic and somatic divisions of the peripheral nervous system

There are two other important differences between the autonomic and somatic divisions. First, most organs under autonomic control receive two sets of nerve fibres – sympathetic fibres and parasympathetic fibres. Thus, the ANS is subdivided into sympathetic and parasympathetic divisions. Second, in the somatic nervous system, the neurotransmitter that carries the message from the neuron to the skeletal muscle is acetylcholine; in the ANS, either acetylcholine or noradrenaline carries the message to the effector. Table 4.3 summarises these and other differences between the autonomic and somatic divisions.

TABLE 4.3 Comparison of the autonomic and somatic divisions of the peripheral nervous system

CHARACTERISTIC	AUTONOMIC DIVISION	SOMATIC DIVISION
Effectors	Heart muscle, involuntary muscle, glands	Skeletal (voluntary) muscles
General function	Adjustment of the internal environment (homeostasis)	Response to the external environment
Efferent (outward) pathways	Two nerve fibres from the CNS to the effector with a synapse in a ganglion	One nerve fibre from the CNS to the effector; no synapse and no ganglion
Neurotransmitter at effector	Acetylcholine or noradrenaline	Acetylcholine
Control	Usually involuntary	Usually voluntary
Nerves to target organ	Two sets – sympathetic and parasympathetic	One set
Effect on target organ	Excitation or inhibition	Always excitation

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Impulses from the sympathetic and parasympathetic divisions have differing effects on organs or tissues. It is not possible to generalise and say that one set of fibres speeds up organ functioning and the other slows it down. However, it can be said that the parasympathetic division generally produces responses that maintain the body during relatively quiet conditions. On the other hand, the sympathetic division tends to produce responses that prepare the body for strenuous physical activity. These responses are often called fight-or-flight responses because they prepare the body for situations that may involve aggression or fleeing from a threat (see below).

The message from the autonomic nerves to the muscles and glands under their control is carried by a neurotransmitter at the nerve endings. Parasympathetic nerve endings release acetylcholine, and sympathetic nerve endings release noradrenaline.

Fight-or-flight response

Under normal circumstances, we are not aware of the activities of the ANS. This does not mean that it is in a resting state. When you are sitting quietly reading a human biology textbook, your sympathetic and parasympathetic nerves are sending out impulses to the internal organs to maintain the stability of the body's functions. For example, the heart has an inbuilt rate of contraction of about 100 beats per minute. While at rest, parasympathetic stimulation keeps this down to around 70 to 80 beats per minute.

FIGURE 4.20 a The sympathetic nervous system produces fight-or-flight responses; **b** The parasympathetic nervous system maintains the body during quiet times

In threatening situations, the balance between sympathetic and parasympathetic stimulation is upset and the sympathetic becomes dominant. Situations that involve fear, anger, stress, danger or competition provoke what is called a **fight-or-flight response** or **alarm reaction**. These responses prepare the body for increased activity (in other animals, to fight or to flee) and, therefore, rely on skeletal muscles producing movement and an increased level of alertness to think and act quickly. This state of preparedness needs a greater supply of oxygen and glucose and, hence, increased blood flow to relevant structures. Therefore, activation of the sympathetic division results in the following responses:

- The rate and force of contraction of the heart increase, with a consequent increase in blood pressure.
- Blood vessels dilate in organs involved in strenuous activity, such as the skeletal muscles, heart and liver.
- Blood vessels constrict in organs not involved in activity, such as the kidney, stomach, intestines and skin.
- Airways in the lungs dilate and the rate and depth of breathing increases.
- Blood glucose level rises, because the liver converts more glycogen into glucose.
- Secretion from sweat glands increases.
- The adrenal medullae release the hormones adrenaline and noradrenaline, which intensify and prolong the above responses.

Table 4.4 summarises the effects of the sympathetic and parasympathetic fibres of the ANS.

TABLE 4.4 Summary of the effects of the autonomic nervous system

STRUCTURE	EFFECT OF SYMPATHETIC STIMULATION	EFFECT OF PARASYMPATHETIC STIMULATION
Heart	Increases rate and strength of contraction	Decreases rate and strength of contraction
Lungs	Dilates bronchioles (fine air passages in the lungs)	Constricts bronchioles
Stomach, intestines	Decreases movement	Increases movement
Liver	Increases breakdown of glycogen and release of glucose	Increases uptake of glucose and synthesis of glycogen
Iris of the eye	Dilates pupil	Constricts pupil
Sweat glands	Increases sweat secretion	No effect
Salivary glands	Decreases secretion of saliva	Increases secretion of saliva
Blood vessels of: • skin • skeletal muscle • internal organs	 Constricts vessels Dilates vessels Constricts vessels (except in heart and lung)	 Little effect No effect Little effect
Urinary bladder	Relaxes muscles of wall	Constricts muscles of wall
Adrenal medulla	Stimulates hormone secretion	No effect

Note: This is only a summary of the most important effects of sympathetic and parasympathetic stimulation. There are many other effects of these divisions.

Key concept

The autonomic nervous system is responsible for maintaining the internal environment of the body. It contains sympathetic and parasympathetic divisions that work together to prepare the body for strenuous activity or rest, respectively.

Activity 4.3
Observing an autonomic reflex

Peripheral nervous system
This website provides more information about the peripheral nervous system.

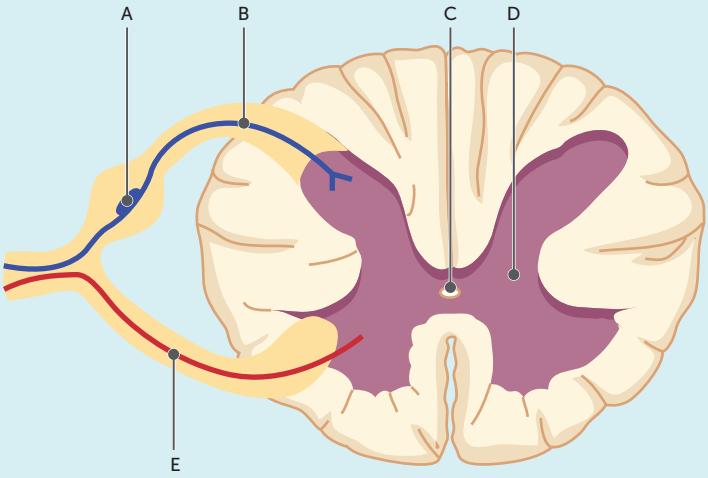
Overview of the autonomic nervous system
This website provides more information on the autonomic nervous system and its disorders.

4.2 Nervous system overview

Questions 4.2

RECALL KNOWLEDGE

1. What structures make up the peripheral nervous system?
2. State the number of:
   - **a** cranial nerves
   - **b** spinal nerves.
3. 'All of the nerves in the peripheral nervous system are mixed nerves, made up of both motor and sensory fibres.' Discuss this statement.
4. Label structures A–E on the diagram below.



5. Draw a tree diagram to show the divisions of the peripheral nervous system.
6. Which division of the PNS takes messages:
   - **a** into the CNS from the internal organs?
   - **b** from the CNS to the muscles and glands?
   - **c** from the CNS to the involuntary muscles?
   - **d** into the CNS from the skin and muscles?
7. Compare and contrast the sympathetic and parasympathetic nervous systems.
8. Explain how the sympathetic nervous system is able to prepare the body for a fight-or-flight response.

APPLY KNOWLEDGE

9. State one similarity and one difference between an ascending tract and a sensory nerve.
10. Suggest what would happen if the body could not produce acetylcholine.

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CHAPTER 4 ACTIVITIES

ACTIVITY 4.1 Examining a dissected brain

A sheep's brain is similar to that of a human, so examining a sheep's brain can help you to understand what a human brain is like.

You will need

Sheep's brain; dissecting instruments; dissecting board or tray; gloves; safety glasses

What to do

1. Place the brain on the dissecting board.
2. Observe the brain from the top.
   - a Describe what the brain looks like.
   - b Identify the cerebrum (both left and right hemispheres) and the cerebellum.
   - c Take note of the convolutions, sulci and fissures.
   - d Take a photo of the brain from the top.
3. Using forceps, peel off some of the membrane that covers the surface of the cerebrum.
   - a This membrane is the inside layer of the meninges. Describe the appearance of the meninge.
   - b Take a photo of the meninge.
4. Turn the brain over and observe it from the bottom.
   - a Describe what the brain looks like.
   - b Identify the cerebellum, brain stem, medulla oblongata and, if possible, the pituitary gland.
   - c Take a photo of the brain from the bottom.
5. Carefully cut the brain in half by cutting between the left and right hemispheres.
   - a Describe what the brain looks like.
   - b Identify where the left and right hemispheres were joined together. This is called the corpus callosum.
   - c Take a photo of the brain.
6. Cut the cerebrum in half by cutting one hemisphere from left to right.
   - a Describe what the cerebrum looks like on the inside.
   - b Identify the white matter (myelinated fibres) and grey matter (cell bodies and unmyelinated fibres).
   - c Predict why the myelinated fibres make up the white matter.
   - d Take a photo of the inside of the cerebrum.
7. Cut the cerebellum in half.
   - a Describe what the cerebellum looks like on the inside.
   - b Take a photo of the inside of the cerebellum.
8. Cut the spinal cord in half.
   - a Describe what the spinal cord looks like on the inside.
   - b Describe how this is the same as or different from the inside of the cerebrum.
   - c Take a photo of the inside of the spinal cord.
9. Feel the texture of the different parts of the brain.
   - a Describe what the brain feels like.
   - b Suggest why it feels like this.

10 Packing up.
- **a** Wrap the brain in newspaper and put it in the bin.
- **b** Wash and wipe your chopping board and put it away.
- **c** Place the scalpel and probe in the containers provided. Make sure they are placed pointy end down.

Putting it together

1 Collate your photos on to one page.
- **a** Label each to show how it was taken (e.g. 'Top of brain').
- **b** Add labels to identify the parts of the brain that you can see in each photo.
- **c** Add labels to identify the lobes of the cerebrum.

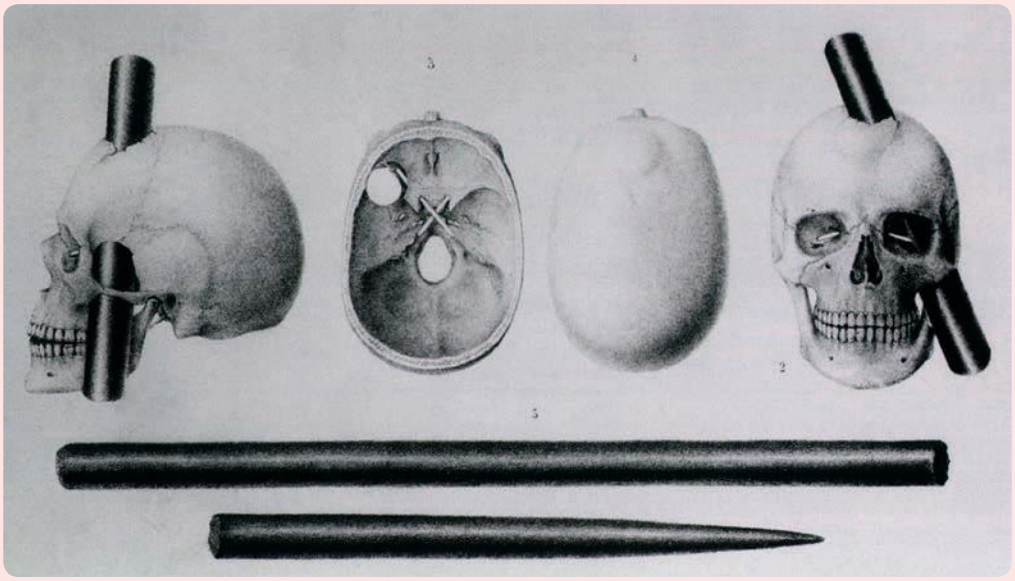
2 A human brain has many more convolutions than a sheep's brain. Explain the significance of the greater number of convolutions in humans.

3 What is the function of the inner meningeal layer?

4 You would have noticed that the inside of the brain is moist.
- **a** What is the name of the fluid that fills spaces inside the brain?
- **b** Where does the fluid come from, and what is its function?

ACTIVITY 4.2 Phineas Gage

In 1848, Phineas Gage was foreman of a gang of workers building a railway in the American state of Vermont. His gang's job was to blast rock with explosives. On 13 September of that year, after placing gunpowder in a hole they had drilled in a rock, Gage began to compact the charge by pushing an iron rod into the hole. The rod was 3 cm in diameter and more than 1 m long. The charge exploded and drove the rod through Gage's skull. The image below shows how it entered the side of his face, passed behind his left eye and exited through the top of his cranium. Reports claimed that the iron rod landed 25 m away. Remarkably, Gage survived the horrendous injury to his brain. He was able to work again (but not with explosives!) and lived for 12 years following the accident.



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After the accident, changes were reported in Gage's behaviour, but his bodily functions such as heartbeat, breathing, digestion, metabolism and regulation of body temperature were unaffected.

Follow the weblink for help in answering the following questions.

1. How is it possible that Gage was able to function relatively normally with damage to such a large and vital part of his brain?
2. Changes in a person's functioning or behaviour as a result of injury to the brain were used by scientists to determine the functions of the affected parts of the brain. Were scientists able to learn anything about the brain from Gage's injury?
3. Did Gage's injury have any positive benefits for medical science?

Phineas Gage
This is a good place to begin your search for answers to these questions.

ACTIVITY 4.3 Observing an autonomic reflex

Work in pairs for this activity, with one person acting as the subject and the other as observer.

The subject should close his or her eyes for at least one minute. While facing a window, or other bright light, the subject then opens their eyes while the observer looks closely to see what happens.

Swap roles and repeat the activity.

1. What change was observed in the subject's pupils when the eyes were opened?
2. What change was observed in the subject's iris when the eyes were opened?
3. Why is the response that you observed described as a reflex?
4. Many reflexes are described as protective. Is the reflex that you observed a protective reflex? Explain.
5. Would it be possible to consciously prevent the response that you observed from occurring?
6. Which division of the autonomic nervous system caused the response that you observed?
7. Optometrists place drops of a drug in the eyes to dilate the pupils so that the eyes can be examined. The drug blocks receptors for acetylcholine. Suggest why such a drug placed in the eyes could cause the pupil to dilate.

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CHAPTER 4 SUMMARY

- The nervous system is made up of the brain and spinal cord (the central nervous system) and nerves (the peripheral nervous system).
- The central nervous system (CNS) processes incoming messages and initiates outgoing messages.
- The CNS is protected by bone (cranium and vertebrae), meninges and cerebrospinal fluid (CSF).
- The meninges are made up of three layers – the dura mater, arachnoid mater and pia mater – between the inside of the bone and the outside of the brain and spinal cord.
- CSF is found between the inner and middle layers of the meninges, as well as in cavities in the brain and the central canal of the spinal cord. It acts as a shock absorber, supports the brain, and transports nutrients and wastes.
- The brain is made up of different structures that carry out different functions.
- The cerebrum makes up the largest portion of the brain. Its outer layer, the cerebral cortex, is made up of grey matter that surrounds the white matter. There is also grey matter deep inside the cerebrum that is called the basal ganglia.
- Grey matter is made up of cell bodies, dendrites and unmyelinated axons, whereas the white matter is made up of myelinated axons. Tracts in the white matter connect different areas of the brain.
- The cerebral cortex is folded, forming convolutions (ridges), sulci (shallow downfolds) and fissures (deeper downfolds).
- The cerebrum can be divided into left and right hemispheres that are joined by the corpus callosum. It can also be divided into five lobes: parietal, frontal, occipital, temporal and insula.
- The cerebral cortex is involved in thinking, reasoning, learning, memory, intelligence, sense of responsibility, movement and senses. These functions allow the cortex to be divided into sensory areas, motor areas and association areas.
- The cerebellum, a folded structure at the base of the brain, is responsible for posture, balance and the coordination of movement.
- The hypothalamus helps maintain a constant internal environment by regulating heart rate, blood pressure, digestion, temperature, water intake and emotions.
- The medulla oblongata contains the cardiac, respiratory and vasomotor centres and plays a role in automatically adjusting body functions.
- The spinal cord runs down the back in the vertebral column. It contains myelinated fibres that take impulses towards the brain in the ascending tracts and away from the brain in the descending tracts.
- The peripheral nervous system (PNS) is made up of the nerves taking messages into and out of the CNS, and groups of nerve cell bodies called ganglia.
- Nerves can be classified as cranial or spinal, based on where they connect with the CNS.
- There are 12 pairs of cranial nerves. Most of these are mixed nerves, containing motor and sensory fibres.
- There are 31 spinal nerves. All of these are mixed nerves. The motor fibres exit the spinal cord via the ventral root, while the sensory fibres enter the spinal cord via the dorsal root. The cell bodies of the motor fibres are located in the dorsal root ganglion.
- The PNS can be divided into afferent and efferent divisions. The afferent division is made up of sensory neurons taking messages from the receptor into the CNS. The efferent division is made up of motor neurons taking messages

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away from the CNS to the muscles and glands.

- The afferent division is further divided into somatic sensory neurons from the skin and muscle, and the visceral sensory neurons from the internal organs.
- The efferent division is further divided into the somatic division, which takes impulses to the skeletal muscles, and the autonomic division, which takes messages to the heart, involuntary muscles and glands.
- The autonomic nervous system operates without conscious control and helps to regulate body functions such as heart rate, blood pressure, body temperature, digestion, air flow and energy release. It is made up of the parasympathetic division, which maintains the body during rest, and the sympathetic division, which prepares the body for strenuous activity.

CHAPTER 4 GLOSSARY

Afferent division Part of the peripheral nervous system containing nerve fibres that carry impulses into the brain and spinal cord; also called sensory division

Alarm reaction *see* fight-or-flight response

Arachnoid mater The middle meningeal layer

Ascending tract Any of the sensory nerve fibres in the central nervous system that carry impulses towards the brain

Association area A part of the cerebral cortex concerned with intellectual and emotional processes such as memory, reasoning, judgement and personality

Autonomic division The part of the efferent division of the peripheral nervous system that carries nerve impulses from the brain and spinal cord to internal organs, involuntary muscles and glands; also called the autonomic nervous system

Autonomic nervous system (ANS) The part of the nervous system that controls the body's internal environment

Basal ganglia The masses of grey matter inside each cerebral hemisphere

Cardiac centre The part of the brain that regulates heartbeat; located in the medulla oblongata

Central canal A hollow that runs through the centre of the spinal cord; filled with cerebrospinal fluid

Central nervous system (CNS) The part of the nervous system that consists of the brain and spinal cord

Cerebellum The part of the brain behind and below the cerebrum; concerned with coordination of movement

Cerebral cortex The outer layer of the cerebrum, made up of grey matter

Cerebral hemisphere One of the two halves of the cerebrum

Cerebrospinal fluid (CSF) Fluid produced in the cavities of the brain; fills the brain cavities and surrounds the brain and spinal cord

Cerebrum The largest part of the brain; made up of left and right hemispheres

Convolution An upward fold of the cerebral cortex of the brain; also called gyrus

Corpus callosum A bundle of nerve fibres that links the two cerebral hemispheres

Cranial nerve One of the 12 pairs of nerves that arise from the brain

Cranium The part of the skull that contains the brain

Descending tract Any of the motor nerve fibres in the central nervous system that carry impulses away from the brain

Dorsal root One of the two roots that link a spinal nerve to the spinal cord; located towards the back of the body and contains axons of sensory neurons

Dorsal root ganglion A group of nerve cell bodies located in the dorsal root of a spinal nerve

Dura mater The outer meningeal layer

Efferent division Part of the peripheral nervous system containing nerve fibres that carry impulses out of the brain and spinal cord; also called motor division

Fight-or-flight response A response preparing the body for increased activity; brought about by stimulation of the sympathetic division of the autonomic nervous system

Fissure A deep downfold in the cerebral cortex of the brain

Frontal lobe One of the five lobes of each cerebral hemisphere

Ganglia Groups of nerve cell bodies outside the brain or spinal cord; singular: ganglion

Grey matter The part of the brain and spinal cord made up of nerve cell bodies and unmyelinated fibres

Gyrus An alternative name for a convolution of the cerebrum; plural: gyri

Hypothalamus The part of the brain lying just below the thalamus; controls

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many homeostatic mechanisms, such as body temperature, water balance and heart rate

Insula A part of the cerebrum that is buried deep inside the brain; considered a fifth lobe of each cerebral hemisphere

Longitudinal fissure The longest fissure in the human brain; almost separates the cerebrum into two halves

Medulla oblongata The part of the brain that joins to the spinal cord

Meninges The membranes covering the brain and spinal cord

Motor area A part of the cerebral cortex that controls muscle movement

Motor fibre A fibre that carries impulses away from the central nervous system

Myelin White, fatty material that surrounds some nerve fibres

Nervous system The system involved with control and coordination of the body

Occipital lobe One of the five lobes of each cerebral hemisphere

Parasympathetic division One of the two divisions of the autonomic nervous system; opposes the function of the sympathetic division; also called parasympathetic nervous system

Parietal lobe One of the five lobes of each cerebral hemisphere

Peripheral nervous system (PNS) The part of the nervous system that connects the central nervous system with the receptors, muscles and glands

Pia mater The inner meningeal layer

Respiratory centre The part of the brain that regulates breathing rate; located in the medulla oblongata

Sensory area A part of the cerebral cortex that interprets impulses from receptors

Sensory fibre A fibre that carries impulses into the central nervous system

Somatic division The part of the efferent division of the peripheral nervous system that carries nerve impulses from the brain and spinal cord to skeletal muscles and skin; also called the somatic nervous system

Somatic sensory neuron A neuron in the afferent division of the peripheral nervous system that takes impulses from the skin and muscles to the central nervous system

Spinal cord The nerve cord that extends from the brain to about waist level; enclosed in the vertebrae

Spinal nerve One of the 31 pairs of nerves that arise from the spinal cord; joined to the spinal cord by dorsal and ventral roots

Sulci Shallow downfolds between convolutions of the cerebral cortex; singular: sulcus

Sympathetic division One of the two divisions of the autonomic nervous system; opposes the function of the parasympathetic division; also called sympathetic nervous system

Temperal lobe One of the five lobes of each cerebral hemisphere

Tract A bundle of nerve fibres in the central nervous system

Vasomotor centre The part of the brain that regulates the diameter of blood vessels; located in the medulla oblongata

Ventral root One of the two roots that link a spinal nerve to the spinal cord; located towards the front of the body; contains axons of motor neurons

Vertebral canal The opening in the vertebrae through which the spinal cord passes

Visceral sensory neuron A neuron in the afferent division of the peripheral nervous system that takes impulses from the internal organs to the central nervous system

White matter The part of the brain and spinal cord made up of myelinated fibres

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CHAPTER 4 REVIEW QUESTIONS

Recall

1 Describe the three structures that protect the central nervous system.

2 a What is cerebrospinal fluid?
b Where does CSF come from?
c Where does CSF go to?
d What does CSF do?

3 a Describe the cerebral cortex.
b List the advantages of the cerebral cortex being folded.
c What is the difference between a sulcus and a fissure?

4 a Describe the functions of the cerebral cortex.
b Name the three types of area in the cerebral cortex and identify the function of each type.

5 a Describe the location of the hypothalamus.
b List some of the functions of the hypothalamus.

6 a Describe the location of the cerebellum.
b What are the main functions of the cerebellum?

7 How many pairs of nerves arise from each of the brain and spinal cord?

8 On what sort of nerve would you find a ventral root and a dorsal root? Explain where these roots are located.

9 Does the autonomic nervous system require conscious control? Why is this important?

10 Describe four differences between the somatic and autonomic divisions of the peripheral nervous system.

11 What is a ganglion?

12 In general terms, what is the difference between responses brought about by the sympathetic and parasympathetic divisions of the autonomic nervous system?

Explain

13 Explain how the structure of the corpus callosum allows it to achieve its function.

14 Explain the medulla oblongata's role in adjusting normal body functions.

15 Explain what a mixed nerve is.

16 a What is the difference between the afferent and efferent divisions of the peripheral nervous system?
b What is the difference between the somatic and autonomic divisions of the efferent division of the peripheral nervous system?

Apply

17 Compare and contrast the grey matter in the cerebrum, cerebellum and spinal cord.

18 After sustaining a head injury in a car accident, a person had difficulty chewing and swallowing. What part of the brain could have been damaged? Justify your answer.

19 Paraplegia (inability to move the legs) may be caused by an injury to the spinal cord. Explain why such an injury could result in paraplegia.

20 A person could survive complete destruction of one of the cerebral hemispheres, which make up nearly 40% of the volume of the brain. By contrast,

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destruction of the hypothalamus, which is only about the size of an almond, would result in certain death. Explain the reasons for this difference.

21 If the ventral root of a spinal nerve were damaged, would it affect the sensory functions or the motor functions of that nerve? Explain.

22 a List four stimuli that could lead to a fight-or-flight response.
b List four responses that would prepare the body for fight or flight.

23 It is sometimes said that the sympathetic division of the autonomic nervous system produces fight-or-flight responses, while the parasympathetic division is concerned with 'rest and digest'. Do you think these are appropriate descriptions for the two divisions? Explain your answer.

24 Urinary retention (inability to empty the bladder or incomplete emptying of the bladder) and incontinence (uncontrollable, involuntary leaking of urine) are both possible symptoms of disease of the autonomic nervous system. Which part of the autonomic division would be affected in each case? Explain your answer.

25 If the dorsal root of a spinal nerve were damaged, would there be any impairment of the autonomic functions controlled by that nerve? Explain your answer.

26 Would a drug that stimulated acetylcholine receptors affect the autonomic division, the somatic division or both? Give reasons for your answer.

27 The drug atropine occupies acetylcholine receptors at the synapse. Ophthalmologists once used atropine when they needed to dilate a patient's pupils. Explain why atropine would have this effect.

Extend

28 In severe cases of epilepsy, as a last resort, the corpus callosum may be severed so that the two cerebral hemispheres can no longer communicate with each other. Patients who have had this procedure are commonly referred to as having a 'split brain'. As each of the two cerebral hemispheres has separate functions, a split brain has a significant impact on the performance of simple tasks. Use references to find out the effects that a split brain would have on a person's functioning.

29 We all need a certain amount of sleep to continue to function normally. Conduct research to find out:
a what happens to the brain during sleep
b the difference between deep sleep and rapid eye movement (REM) sleep
c the difference between sleep and a coma.

30 Lie detectors measure ANS activity. Their use is based on the idea that when a person lies, there are involuntary changes in their body functions. Find out:
a what kinds of things lie detectors measure and how they are measured
b whether it is the activity of the sympathetic or parasympathetic divisions that produces the responses measured
c how reliable lie detectors are, and reasons for their reliability or unreliability.

31 As people get older, changes occur in the nervous system. Some changes are serious enough to be called a disease; an example is Alzheimer's disease. Other changes are just a natural part of ageing. Conduct research to find out:
a the changes to the nervous system that occur in everyone as they get older
b the reasons for those changes
c what can be done to reduce or delay the changes to the nervous system.

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5 HOMEOSTASIS CONTROLS BLOOD GLUCOSE AND BODY TEMPERATURE

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including the collection of data related to homeostasis and the use of models of disease transmission, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions.
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

SCIENCE UNDERSTANDING

Homeostasis

- homeostatic processes involve nerves and hormones in maintaining the body's internal environment within tolerance limits through the control of metabolism and physiological and behavioural activities
- thermoregulation occurs by the control of heat exchange and metabolic activity through physiological and behavioural mechanisms
- blood sugar levels are maintained by controlling of sugar uptake, its storage and release by cells and use in metabolism; these processes involve the hormones of the pancreas and adrenal glands

Source: School Curriculum and Standards Authority, Government of Western Australia

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5.1 HOMEOSTASIS

Think of the last time you ran to catch a bus or a train, or ran up several flights of stairs. After such vigorous activity you may have been sweating, your face may have been red, you would have been breathing heavily, and you would have been able to feel your heart beating forcefully and rapidly. All these responses would have occurred automatically, without any conscious thought on your part. Such responses are a part of homeostasis. **Homeostasis** is the process of keeping the environment inside the body fairly constant.

Our body cells work best at a particular temperature, when surrounded by fluid with a particular pH, when given a constant supply of oxygen and glucose, and when wastes are constantly removed. Maintaining these, and other optimum conditions for cell functioning, is all part of homeostasis.

Homeostatic mechanisms help us to be independent of our external environment. For example, if you suddenly plunge into a cold swimming pool, the cells of your brain, liver, stomach, heart and other internal organs will continue to function normally despite the sudden change in external temperature.

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FIGURE 5.1 Homeostasis makes us relatively independent of the external environment

The body's cells are surrounded by fluid, the composition and temperature of which must be maintained within very narrow limits. The important aspects of the internal environment that the body needs to regulate include:

- core body temperature
- pH and concentrations of dissolved substances in the body fluids
- concentration of glucose in the blood
- concentration of oxygen and carbon dioxide in the blood and other body fluids
- blood pressure
- concentration of metabolic wastes.

The maintenance of this **steady state** does not mean that nothing changes. Instead, there is a dynamic equilibrium in which the input and output of materials and energy are balanced. All the systems of the body contribute to homeostasis, not only to supply the cells' needs, but also to maintain a constant cellular environment.

To maintain homeostasis, the body must be able to both sense changes in the internal and external environment and compensate for those changes. The nervous and endocrine systems are the main sensory and controlling body systems; in the case of homeostasis, they operate through feedback systems, many of which involve negative feedback.

Feedback systems

A **feedback system** is a circular situation in which the body responds to a change, or **stimulus**, with the response altering the original stimulus and thus providing feedback. Feedback systems can be negative or positive, depending on whether the response decreases or increases the original stimulus. For example, when we exercise our muscles use glucose to release the energy required for muscle contraction. The muscles absorb glucose from the blood and, consequently, the blood glucose level tends to fall. This is the stimulus. The liver responds by releasing more glucose into the blood. Thus, the response has caused the blood glucose level to go up, which is the *opposite* of the fall in glucose that initiated the response. In this way, the blood glucose level is maintained within a range that is acceptable for efficient cellular functioning.

Feedback systems have a number of common features.

- The *stimulus* is the change in the environment that causes the system to operate.
- The *receptor* detects the change.
- The ***modulator*** is a control centre responsible for processing information received from the receptor and for sending information to the effector.

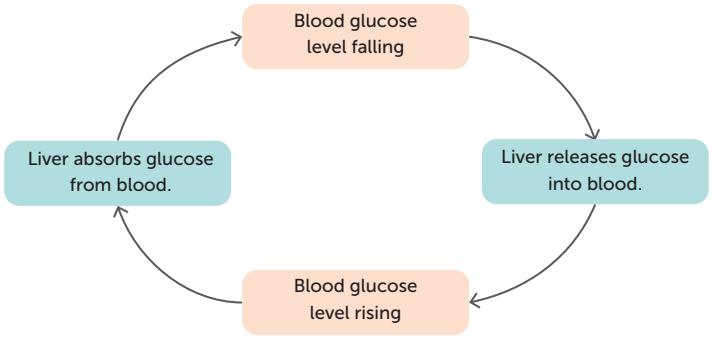
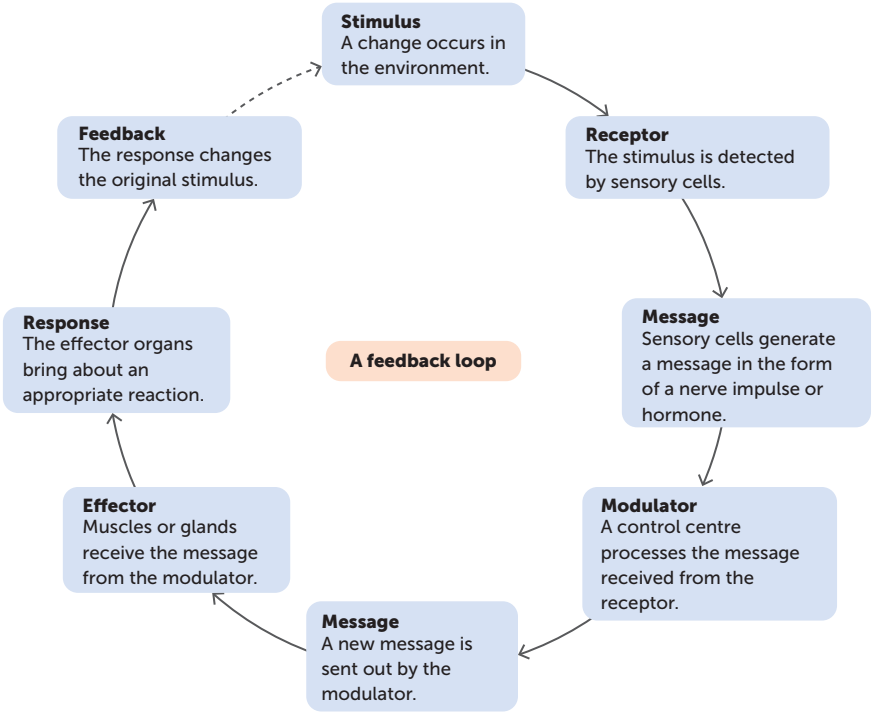


FIGURE 5.2 Example of a feedback system: the feedback loop controlling the body's blood glucose level

- The *effector* carries out a response counteracting or enhancing the effect of the stimulus.
- *Feedback* is achieved because the original stimulus has been changed by the response.

In science, a **model** is a simplified representation of something that is fairly complex. The stimulus–response–feedback model is a simple way of explaining how homeostatic mechanisms work.

FIGURE 5.3 The stimulus–response–feedback model



Key concept

A feedback loop involves a stimulus, receptor, modulator, effector, response and feedback.

Homeostasis and feedback loops
This website has more information about homeostasis and feedback loops.

Homeostatic mechanisms are controlled by both the nervous system and the endocrine system. Both systems detect when the body is beginning to deviate from its normal balanced state: the nervous system sends electrical messages to the appropriate organs so that the change is counteracted; the glands of the endocrine system secrete chemical messengers, or hormones, into the blood. Hormones, however, generally work more slowly than nerve impulses in coordinating homeostasis.

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Negative feedback

In homeostatic mechanisms the response has the effect of reducing or eliminating the stimulus that caused it. This is called **negative feedback**. For example, if you feel cold your response to the stimulus might be to put on a jumper. This response reduces or eliminates the original stimulus of feeling cold. Negative feedback systems are also called **steady state control systems**, as they return the body to a steady state.

Negative feedback
This website has a simple quiz on negative feedback. Test yourself!

An analogy that is often used to explain a negative feedback loop is that of an air-conditioning system in a room or building. Suppose that the thermostat is set to a comfortable 22°C. On a warm day, when the temperature rises above 22°C, the thermostat automatically switches the air conditioner on so that cool air is brought into the room. The air in the room becomes cooler and the thermostat then switches the air conditioner off when it reaches its target temperature. This process repeats itself so that the air temperature remains relatively constant.

Figure 5.4 shows how the components of the air-conditioning system correspond to the components of a negative feedback loop. Note that the temperature fluctuates above and below the temperature set on the thermostat. It is the same with the human body. Things such as the concentration of blood glucose and body temperature fluctuate around a normal level. This fluctuation is called a **dynamic equilibrium**. The point around which conditions fluctuate is called the **set point**. For the air conditioner in our scenario, the set point is 22°C; for human body temperature, the set point is 37°C. **Tolerance limits** are the upper and lower limits between which the levels fluctuate. Within these limits the body functions normally; however, a rise above, or a fall below, them means that the individual's tolerance limits have been exceeded and dysfunctions will occur.

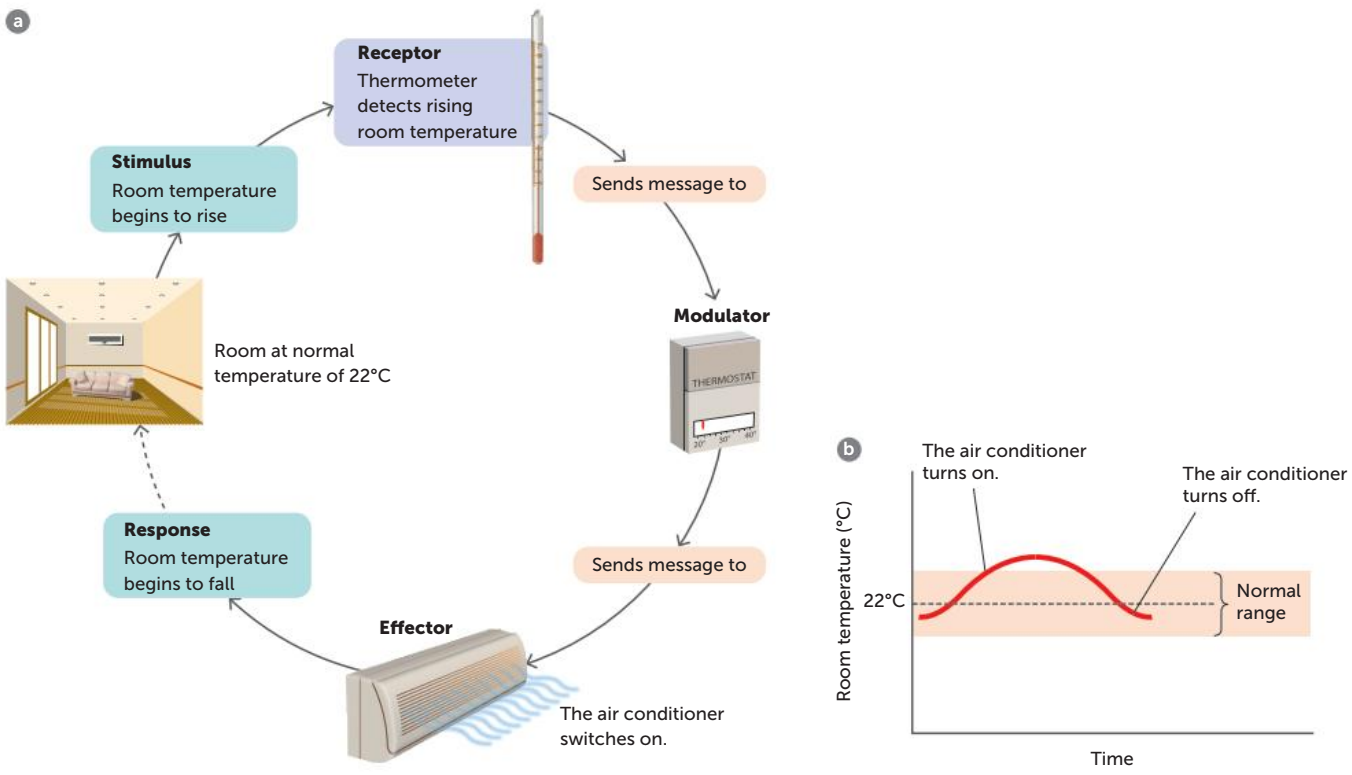


FIGURE 5.4 a An air-conditioning system uses negative feedback to keep the room temperature fairly constant. **b** Room temperature fluctuates around the temperature determined by the thermostat

Key concept

Negative feedback systems maintain homeostasis by producing a response that is the opposite of the original change.

Positive feedback

Positive feedback has no role in homeostasis, but it is included here so that you can understand the difference between positive and negative feedback. When **positive feedback** occurs, the response to a stimulus reinforces and intensifies the stimulus. The intensified stimulus results in an even greater response, and so on. Obviously, this could not result in homeostasis, but there are a few situations in which it does occur in the human body where it is important in controlling processes that must be completed quickly.

An example of positive feedback occurs during childbirth, a process that must be completed as rapidly as possible to avoid stress and injury to mother and baby. Labour is initiated by the secretion of the hormone oxytocin from the posterior lobe of the pituitary gland. Oxytocin causes contractions of the uterus. The contractions push the baby's head against the mother's cervix. Stimulation of the cervix causes it to send nerve impulses to the brain, which responds by instructing the pituitary to secrete more oxytocin. The increased oxytocin makes the uterus contract more strongly. These contractions push the baby's head more forcibly against the cervix, which sends even more impulses to the brain, and so the uterine contractions are increasingly intensified. Once the baby is delivered, the cervix is no longer stretched; it ceases sending nerve impulses to the brain and the positive feedback cycle stops.

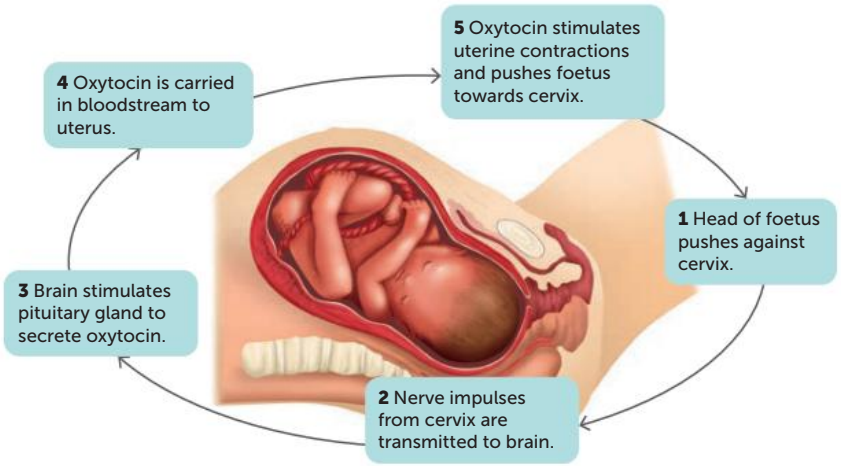


FIGURE 5.5 Childbirth is regulated by a positive feedback system

Another example of positive feedback is blood clotting. This process must be completed quickly to minimise blood loss.

However, positive feedback can sometimes be harmful. An example is a high fever. A small rise in body temperature can be beneficial in fighting infection; however, if body temperature rises above 42°C, a dangerous positive feedback loop can occur. The raised body temperature causes a higher metabolic rate that produces more heat, which raises the temperature still further. This increases the metabolic rate and so the temperature spirals upwards. Unless medical treatment is given, death will result when body temperature reaches about 45°C.

WS

5.1 Homeostasis

Questions 5.1

RECALL KNOWLEDGE

1 Define 'homeostasis'.
2 Fill in the blanks for the negative feedback loop.
Stimulus → _____ → _____ → _____ → _____ → _____ → feedback
3 List four characteristics that are maintained by homeostasis.
4 Describe two positive feedback systems.
5 Use a flow chart to model the feedback loop when drinking water maintains fluid levels in the body on a very hot day.

APPLY KNOWLEDGE

6 Explain the difference between the set point and tolerance limit.
7 Explain why a positive feedback system would not achieve homeostasis.

5.2 REGULATION OF BLOOD SUGAR

Sugar in the blood is in the form of glucose. When we talk about 'blood sugar', we are really talking about the amount of glucose in the blood. All cells need a constant supply of glucose because it is the source of energy for all the cells' activities, such as movement, reproduction, synthesising molecules, active transport and many others. Energy is released from glucose molecules by cellular respiration.

Glucose + oxygen → carbon dioxide + water + energy

The body's source of glucose is the food we eat. Carbohydrates in our food are broken down into glucose during digestion and then absorbed into the blood through the walls of the small intestine. After a meal, blood glucose concentration can rise sharply. Homeostatic mechanisms then begin to operate to reduce the blood glucose concentration and maintain it at the normal level. Any excess glucose in the blood must be removed and stored ready for use in cellular activities between meals.

Science Photo Library/John Birdsall Social Issues Photo Library

FIGURE 5.6
A glucometer (blood glucose meter) being used to measure the level of glucose in the blood. A small sample of blood is taken from a prick in the patient's finger, and placed on the white strip of the glucometer. This patient's reading of 10.4 millimoles/litre (mmol/L) is equivalent to 187mg of glucose per decilitre of blood (mg/dl)

Glucose and glycogen

Glucose is stored as **glycogen**, a molecule made of long chains of glucose molecules. The body is able to store about 500g of glycogen: about 100g is stored in the liver and the remainder in skeletal muscle cells. Excess glucose in the is converted to glycogen for storage. Alternatively, when there is not enough glucose in the blood, some of the glycogen can be converted to glucose.

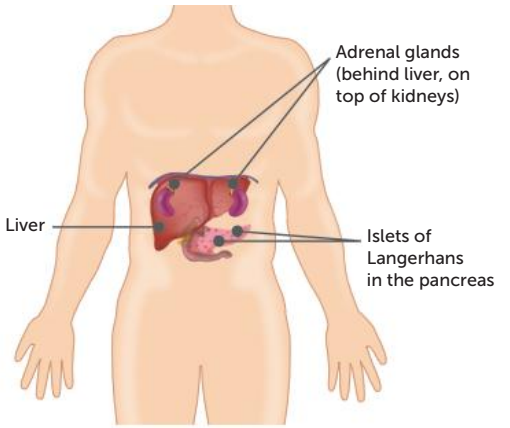


FIGURE 5.7 The main organs involved in the regulation of blood glucose. The pancreas and adrenal glands secrete hormones that affect the level of glucose in the blood, while the liver stores excess glucose as glycogen

FIGURE 5.8
A summary of glucose–glycogen conversions

Role of the liver

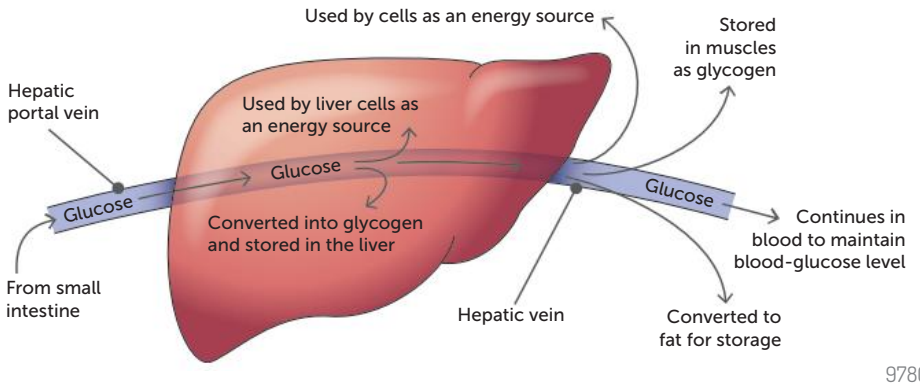
The liver is in the upper part of the abdominal cavity just below the diaphragm. It is the largest gland in the body and has a very important role in the control of blood sugar concentration. The liver is able to convert glucose into glycogen for storage, or glycogen to glucose for release into the blood.

Most of the liver's blood supply comes through the hepatic portal vein, which brings blood directly from the stomach, spleen, pancreas, and small and large intestines. Thus, the liver has the first chance to absorb the nutrients from digested food.

After a typical meal containing a high proportion of carbohydrates is consumed, the breakdown products, mainly glucose, are absorbed into the blood capillaries of the villi of the small intestine. The hepatic portal vein carries the glucose to the liver, where a number of things may occur. Glucose may:

- be removed from the blood by the liver to provide energy for liver functioning
- be removed by the liver and/or muscles and converted into glycogen for storage
- continue to circulate in the blood, available for body cells to absorb and use as a source of energy
- be converted into fat for long-term storage if it is in excess of that required to maintain both normal blood sugar and tissue glycogen levels.

FIGURE 5.9
The fate of glucose absorbed in the small intestine



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Glucose molecules are chemically joined in long chains to form glycogen molecules. This process, known as **glycogenesis**, is stimulated by the pancreatic hormone **insulin**. Glycogen itself cannot be used by cells; it must be converted back into glucose or to other simple sugars. Glycogen stored in the liver is available for conversion into glucose to maintain blood sugar levels and supply energy for liver activity. Glycogen in muscle cells provides the glucose required for muscle activity.

If the level of glucose in the blood drops below normal, the glycogen stored in the liver and muscle cells can be broken down into glucose. This process of converting glycogen back into glucose is called **glycogenolysis**. Most frequently, it occurs between meals and is stimulated by another pancreatic hormone, **glucagon**.

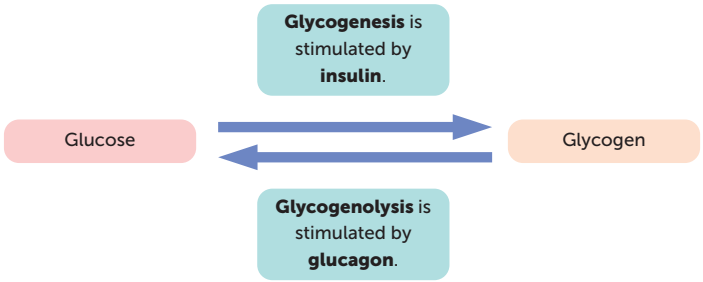

FIGURE 5.10 Glucose–glycogen conversions stimulated by pancreatic hormones

Glycogen stored in the liver is a short-term energy supply. It can provide glucose for body cell use for only about six hours if no other supply is available. If more energy is required, the body uses the energy reserves in stored fat.

TABLE 5.1 Terms relating to glucose metabolism

TERM	MEANING
Glycogenesis	Formation of glycogen from other carbohydrates, especially glucose (in Greek, *genesis* means 'origin' or 'creation')
Glycogenolysis	Breakdown of glycogen to glucose (in Greek, *lysis* means 'to separate or break down')
Gluconeogenesis	Conversion of fats or proteins into glucose (in Greek, *neo* means 'new')

Role of the pancreas

The pancreas is a pale grey gland, 12–15 cm long, lying partly in the curve of the duodenum. Within the pancreas are clusters of hormone-secreting cells called the **islets of Langerhans**. The cells in the islets are of two types:

- **alpha cells** that secrete glucagon
- **beta cells** that secrete insulin.

Insulin from the beta cells causes a *decrease* in blood glucose levels. It does so by:

- accelerating the transport of glucose from the blood into body cells, especially those of the skeletal muscles
- accelerating the conversion of glucose into glycogen in the liver and skeletal muscle (glycogenesis)
- stimulating the conversion of glucose into protein (protein synthesis)
- stimulating the conversion of glucose into fat (lipids) in adipose tissue, or fat storage tissue, a process called **lipogenesis**.

All these activities decrease blood glucose levels.

The level of blood glucose regulates the secretion of insulin via a negative feedback system. When blood glucose levels rise above normal, chemoreceptors in the beta cells of the islets of Langerhans stimulate those cells to secrete insulin. As the level of blood glucose decreases, the cells are no longer stimulated and production is reduced.

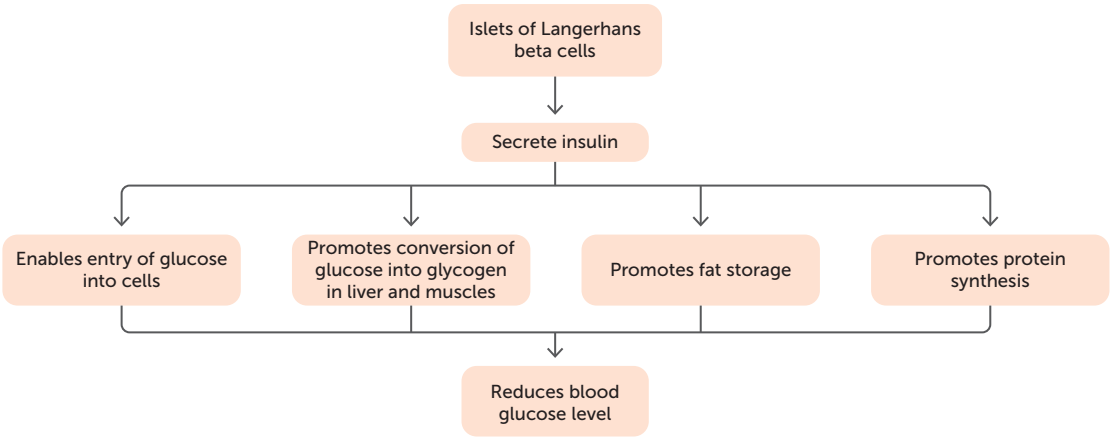

FIGURE 5.11 Effects of insulin on blood glucose levels

Glucagon from the alpha cells causes an *increase* in blood glucose levels. It does this by:

- stimulating glycogenolysis, the conversion of glycogen into glucose, in the liver
- stimulating **gluconeogenesis**, the production of new sugar molecules from fats (lipids) and amino acids, in the liver. This involves the breakdown of lipids in a process called **lipolysis**
- having a mild stimulating effect on protein breakdown.

The glucose formed is released into the blood, and the blood glucose level rises.

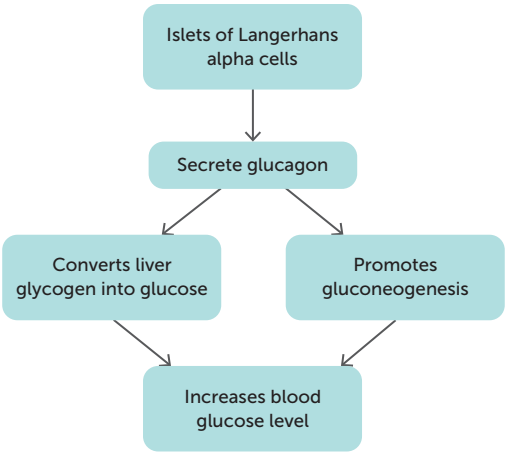

FIGURE 5.12 Effects of glucagon on blood glucose levels

The regulation of the secretion of glucagon, like that of insulin secretion, is directly determined by the level of glucose in the blood and is controlled by a negative feedback system. When the blood glucose falls below normal, chemoreceptors in the alpha cells of the islets of Langerhans stimulate those cells to secrete glucagon. As the blood glucose level increases, the cells are no longer stimulated and production is reduced.

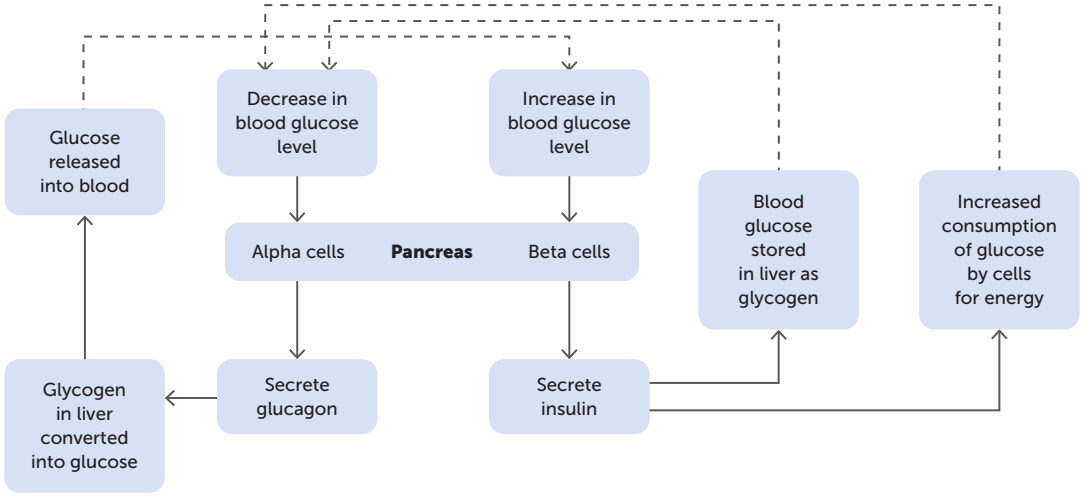

FIGURE 5.13 Regulation of blood glucose by insulin and glucagon

Insulin and diabetes
This website provides more information on the role of insulin and glucagon in blood sugar regulation.

Blood sugar regulation
This website provides more information on the regulation of blood sugar.

Role of the adrenal glands

The adrenal glands are situated just above the kidneys, one gland above each. Each gland is composed of two distinct parts: the outer part is called the **cortex** and the inner part is the **medulla**. The adrenal glands produce a number of hormones, but in discussing the control of blood glucose, we are interested only in the secretion of glucocorticoids by the adrenal cortex, and the secretion of adrenaline (epinephrine) and noradrenaline (norepinephrine) by the adrenal medulla.

The adrenal cortex is stimulated to secrete its hormones by adrenocorticotropic hormone (ACTH) from the anterior lobe of the pituitary gland. The hormones secreted are glucocorticoids, the best known of which is **cortisol**. They regulate carbohydrate metabolism by ensuring that enough energy is provided to the cells. In doing so, they stimulate the conversion of glycogen into glucose during glycogenolysis. They also increase the rate at which amino acids are removed from cells, mainly muscle cells, and transported to the liver. Some of these amino acids may be converted into glucose by the liver during gluconeogenesis if glycogen and fat levels are low.

Glucocorticoids also promote the mobilisation of fatty acids from adipose tissue, allowing muscle cells to shift from using glucose to fatty acids for much of their metabolic energy.

The adrenal medulla synthesises adrenaline and noradrenaline, hormones that produce the same effects as those brought about by sympathetic nerves of the autonomic nervous system. One such effect is the increase of blood glucose levels. In particular, adrenaline elevates blood glucose levels through glycogenolysis and in doing so counteracts the effects of insulin. It stimulates the production of lactic acid from glycogen in muscle cells, which can then be used by the liver to manufacture glucose.

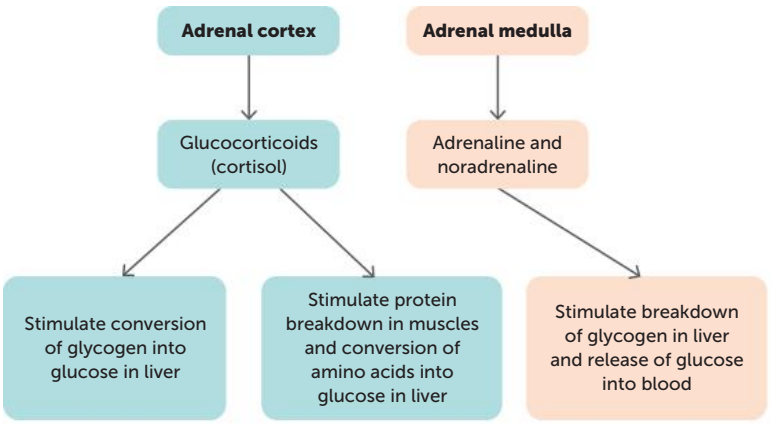

FIGURE 5.14
Effect of adrenal hormones on blood glucose levels

Blood glucose homeostasis

The normal level of glucose in the blood is between 4 and 6 millimoles per litre (5 mmol/L is equivalent to 90 mg/100 mL). Many activities take place to maintain the level within these narrow limits. Our discussion has covered the involvement of the liver, pancreas and adrenal glands. Homeostasis relies on the contribution of all three, working in an integrated manner. Figure 5.15 summarises the main influences on these processes.

WS
5.2 Homeostasis of blood sugar

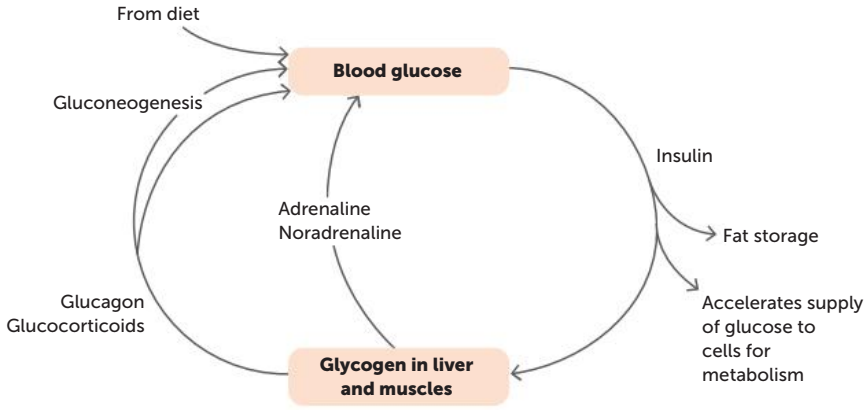

FIGURE 5.15
A summary of blood glucose homeostasis

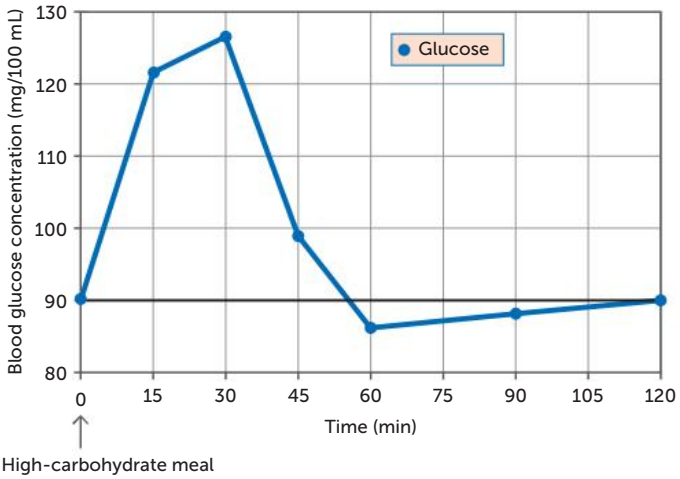

FIGURE 5.16
Change in blood glucose concentration over time following a high-carbohydrate meal

Questions 5.2

RECALL KNOWLEDGE

1. List the organs that play key roles in glucose homeostasis.
2. Define 'glycogenesis', 'glycogenolysis' and 'gluconeogenesis'.
3. Describe what happens to glucose from ingestion to it passing through the liver.
4. Blood glucose levels increase after eating carbohydrates.
   - a. What is the key hormone that will initially be released?
   - b. What organ produces this hormone?
   - c. What cells within the organ produce this hormone?
   - d. What responses does the hormone lead to?
5. Describe the role of the adrenal glands in controlling the level of glucose in the blood.

APPLY KNOWLEDGE

6. Write a word equation for cellular respiration and explain its significance in relation to glucose homeostasis.
7. Use the stimulus–response–feedback model to summarise how the pancreas responds after strenuous activity.

5.3 THERMOREGULATION

On a hot day you would probably get out of the sun, remove some of your outer clothing, and turn on a fan or air conditioner. These are behavioural responses to the high temperature, but at the same time changes would be occurring inside your body that you are not consciously aware of. All these responses, conscious and unconscious, work together to keep the body's core temperature within its tolerance limits. This regulation of body temperature – **thermoregulation** – makes us relatively independent of the environmental temperature.

Heat gain and heat loss

The body temperature of humans remains remarkably constant at about 36.8°C. This is an important aspect of homeostasis because the chemical reactions occurring in cells are very heat sensitive. A temperature of around 37°C is optimum for cellular reactions, and so cells maintained at this temperature function in a stable manner. To achieve this temperature, the heat gained by the body must exactly equal the heat lost from the body. Maintaining this balance is thermoregulation.

Under most conditions, the internal body temperature is higher than the surrounding environmental temperature. The heat produced from metabolic activity helps to maintain this higher level. However, during exercise and other strenuous activity, the increase in metabolic rate generates more heat than the body needs to keep its temperature constant. The excess heat needs to be removed or the body temperature will rise. Increased body temperature can cause nerve malfunction, change in the structure of proteins, and death. Therefore, it is extremely important for the body to be able to regulate its internal temperature and maintain it within fairly precise limits.

Science Photo Library/Tony McConnell

FIGURE 5.17 Thermogram of a ballerina showing relative temperatures at the surface of the body. The warmest areas are white, followed by red, yellow, green and blue, with purple being the coldest

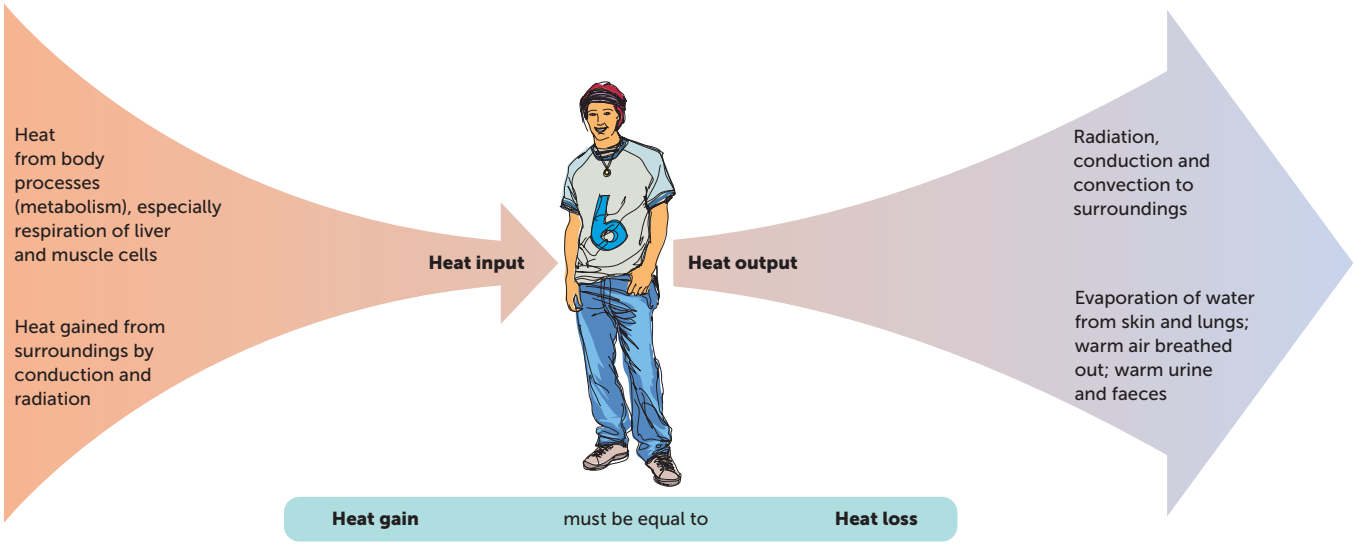

FIGURE 5.18 A constant human body temperature is achieved by a balance between heat gain and heat loss

Although body temperature remains fairly constant, variation does occur as a matter of course. This variation may result from activity or changes in external temperature. In addition, there is a characteristic daily body temperature cycle, with the lowest temperature generally occurring in the morning and the highest in the evening. Also, women have higher temperatures during the second half of the menstrual cycle as a result of the effects of the hormone progesterone.

Heat production

The carbohydrates, proteins and lipids that we eat contain energy in chemical bonds that hold the various parts of the molecule together. This energy is released when the food is oxidised during cellular respiration.

Some of the energy is used for muscle contraction, for active transport of substances across the cell membrane, for building up new complex molecules, and so on. Most of the energy, however, is released in the form of heat.

The rate at which energy is released by the breakdown of food is called the **metabolic rate**. Many factors, such as exercise, stress and body temperature, affect the metabolic rate of an individual. The factor with the greatest effect is exercise. During exercise, muscular activity can increase metabolic rate by up to 40 times, so very large quantities of heat are released.

Metabolic rate also increases in times of stress because of the activities of the autonomic division of the nervous system. Stimulation of sympathetic nerves releases noradrenaline from the nerve endings; noradrenaline increases the metabolic activity of cells. Strong sympathetic stimulation may cause dramatic increases in the metabolic rate, but usually for only a few minutes.

Rising body temperature is another factor that increases metabolic activity. For each 1°C rise in temperature, the rate of biochemical reactions increases by about 10%. Therefore, when an individual is suffering from a high fever, the metabolic rate may be up to double the normal rate.

Regulating body temperature

Temperature receptors

The body has temperature receptors, or **thermoreceptors**. Those in the skin and in some mucous membranes are called **peripheral thermoreceptors.** They detect temperature changes in the external environment and send this information to the hypothalamus. Others are located in the hypothalamus and are called **central thermoreceptors**. These receptors detect the temperature of the internal environment. There are additional thermoreceptors at various internal locations such as the spinal cord and the abdominal organs that provide the hypothalamus with information about the temperature of the internal environment.

There are two types of thermoreceptors: **cold receptors** are stimulated by temperatures lower than normal; and **heat receptors** detect temperatures higher than normal. When cold receptors are stimulated, the hypothalamus receives the information and initiates heat conservation and heat production mechanisms. If heat receptors are stimulated, mechanisms operate to reduce heat production and increase heat loss.

The skin and temperature regulation

The large surface area and location of the skin between the internal and external environment means that it is a very important organ in regulating body temperature. Heat can be lost from the skin by conduction, convection, radiation and evaporation. Therefore, changes in the skin can speed up or slow down the rate at which heat is lost from the body.

- **Conduction** is the transfer of heat by direct contact between particles.
- **Convection** is the transfer of heat by the movement of a liquid or a gas.
- **Radiation** is the transfer of heat by infrared radiation being emitted by objects.
- **Evaporation** is the process of a liquid forming a gas, which absorbs heat energy.

Conduction, convection and radiation
This website explains conduction, convection and radiation in more detail.

Key concept

Heat is lost by conduction, convection, radiation and evaporation.

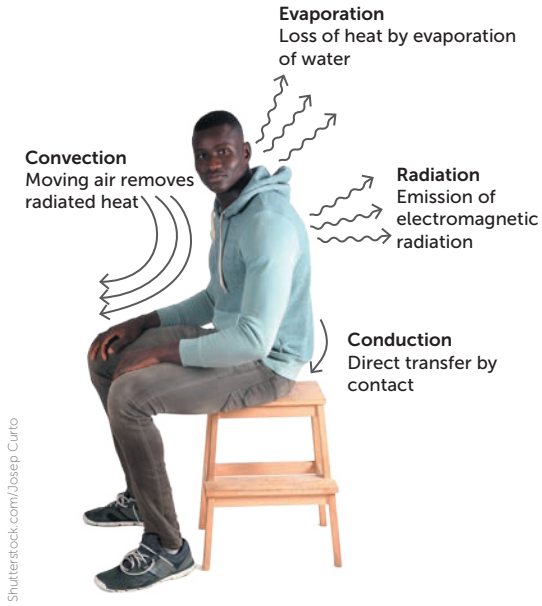

FIGURE 5.19
Methods of heat loss from the body

Blood vessels and heat loss

Blood vessels located in the dermis of the skin carry heat to the skin from the core of the body. The diameter of these arterioles is controlled by autonomic nerves. If the diameter is increased by **vasodilation**, more blood is transported to the capillaries in the skin and the rate of heat loss increases. Alternatively, if the diameter is reduced by **vasoconstriction**, less blood is transported to the capillaries in the skin and the rate of heat loss decreases.

Sweating and heat loss

When large amounts of body heat must be lost and skin blood vessels are already at maximum dilation, sweating must occur. **Sweating** is the active secretion of fluid by the **sweat glands** and the periodic contraction of cells surrounding the ducts to pump the **sweat** to the skin surface. The production and transport of sweat to the skin surface is stimulated by sympathetic nerves.

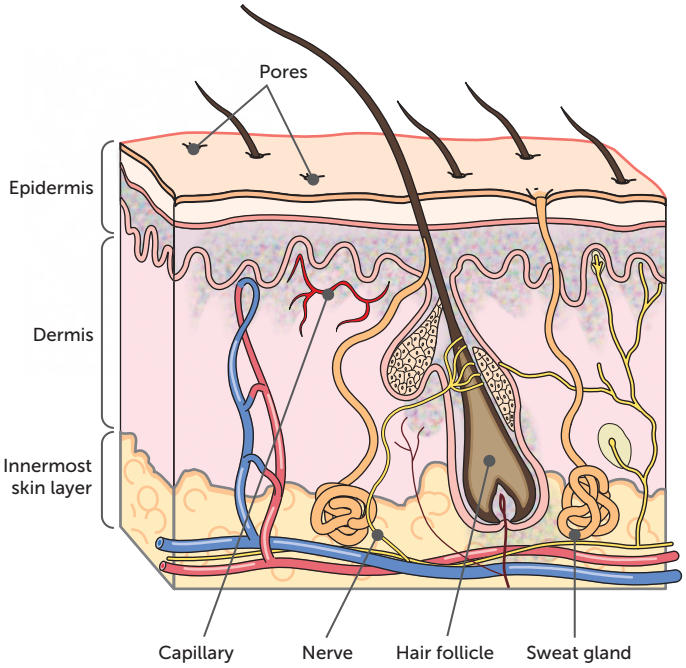

FIGURE 5.20
Section of skin showing sweat glands

Heat transfer
This website provides more information about the different methods of heat transfer: conduction, convection and radiation.

More on heat transfer
This website provides an animation of the various methods of heat transfer.

Sweat is water containing dissolved substances, primarily sodium chloride along with small amounts of urea, lactic acid and potassium ions. Evaporation of sweat from the skin has a cooling effect: heat is removed from the skin when liquid sweat changes into vapour. Cooling of the skin results in cooling of the blood flowing through the skin.

Even in the absence of sweating there is continual loss of water by evaporation from external body surfaces. This evaporation, along with water that is evaporated from the lungs and respiratory passages, accounts for a considerable proportion of the daily heat loss from the body.

Shivering and heat gain

Shivering is due to an increase in skeletal muscle tone, producing rhythmic muscle tremors that occur at a rate of around 10 to 20 per second. As no work is being done, the heat produced by the muscles is released as heat.

Preventing body temperature from falling

If the environmental temperature falls, or if a person goes from a warm room into a cold environment outside, the cold receptors in the skin send messages to the hypothalamus. The hypothalamus then sends out impulses aimed at reducing heat loss and increasing heat production so that body temperature is maintained. The body can respond by making physiological changes (changes in body functioning) and behavioural changes.

- Impulses from the hypothalamus stimulate sympathetic nerves that cause arterioles in the skin to constrict. This vasoconstriction decreases the flow of warm blood to the skin from the internal organs, thus decreasing the transfer of heat from the internal body organs to the skin. The skin becomes cooler because there is less warm blood flowing through it. Less heat will then be lost from the body surface. In this way, vasoconstriction of skin arterioles helps maintain body temperature in cold conditions.

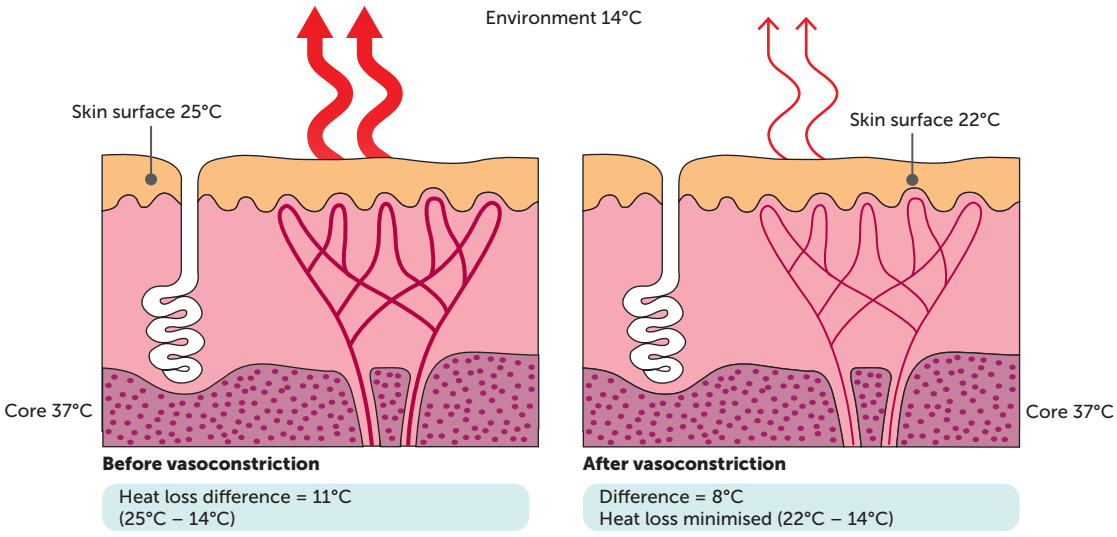

FIGURE 5.21 The effect of vasoconstriction in the skin

- A second response initiated by the hypothalamus is the stimulation of the adrenal medulla by sympathetic nerves. This stimulation results in the medulla secreting adrenaline and noradrenaline into the blood. These hormones bring about an increase in cellular metabolism that leads to an increase in heat production. This helps to maintain internal body temperature in conditions where there is rapid heat loss.

- A fall in body temperature causes the hypothalamus to send stimuli to the parts of the brain that cause shivering, which can increase body heat production several-fold within minutes. Shivering is under the primary control of the hypothalamus, but conscious input from the cerebral cortex can suppress the urge to shiver.
- The fourth response to a fall in body temperature is an increase in the production of thyroxine. The hypothalamus is able to cause the anterior lobe of the pituitary to secrete thyroid-stimulating hormone (TSH), which causes the thyroid gland to release thyroxine into the blood. The increased levels of thyroxine increase the metabolic rate, resulting in an increase in body temperature. This response is slower to have an effect, but it is longer lasting than other responses. The small change in metabolic rate that occurs between the warm summer months and cool winter ones is a result of this response.
- A behavioural response may occur if we become consciously aware of cold conditions. If we feel cold, we can behave in a way that reduces heat loss, such as by putting on an extra jumper or sheltering from a cold wind. Another behavioural response that helps to reduce heat loss is reducing the surface area of the body from which heat can be lost. You may have noticed that when you are cold in bed you tend to curl up into a ball.

Preventing body temperature from rising

When the weather is warm, or when we exercise, the heat produced by metabolism is greater than that needed to maintain a constant body temperature. This excess heat needs to be lost from the body, otherwise the core temperature will rise. Most heat loss occurs through the skin, although smaller amounts of heat are lost with the gases breathed out from the lungs, and with the faeces and urine.

The following responses ensure that body temperature does not rise.

- Vasodilation of skin arterioles increases blood flow through the skin. The skin becomes reddish in colour, surface temperature rises, and there is greater heat loss through radiation and convection.
- When environmental temperatures are above about 28°C, sweating is needed to increase heat loss from the body, as shown in Figure 5.22. The cooling effect of sweating is effective only in environments that are fairly dry. If the air is very humid, sweat cannot evaporate and, therefore, does not absorb heat from the body. If both humidity (the water vapour concentration of the air) and temperature are high, individuals often suffer considerable discomfort as the sweat remains

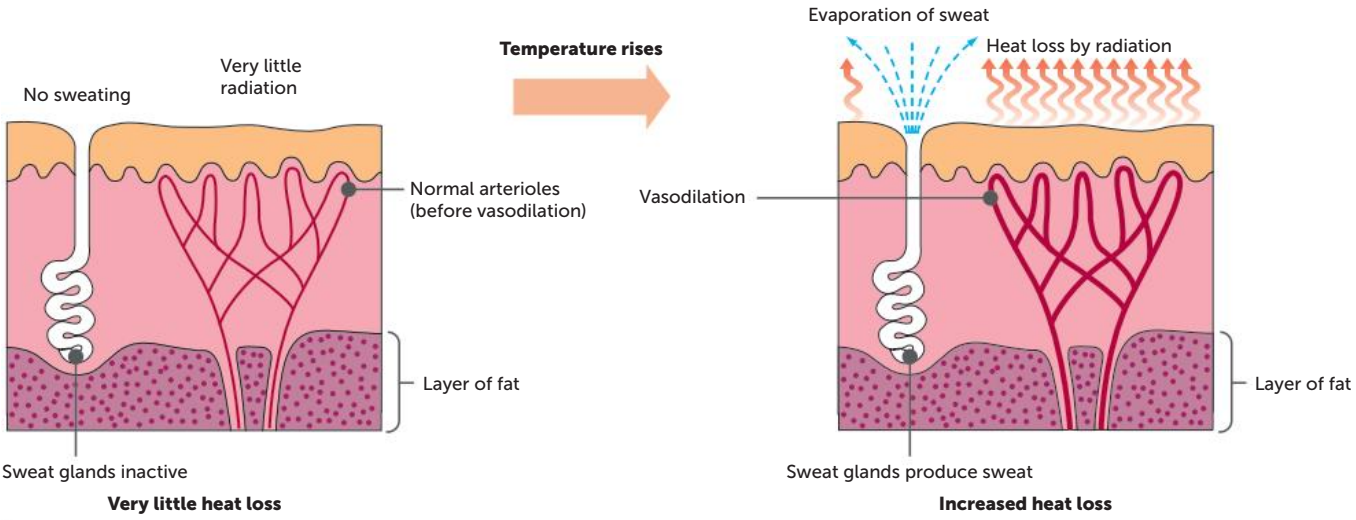

FIGURE 5.22 Heat loss from the skin can be increased by vasodilation of skin blood vessels and by sweating

on the skin or simply drips off. It is only in a low-humidity environment that sweating is a really effective means of preventing body temperature from rising. If environmental temperatures are greater than 37°C, heat is gained from the environment and the evaporation of sweat is then the only avenue for heat loss.

- In the long term there can be a decrease in metabolic rate, which means less heat is produced in the body. Such a decrease is brought about by reduction in the secretion of thyroxine, a response that occurs in summer when there is much less heat loss than in winter.
- Behavioural responses can also help to prevent the body temperature rising. Actions such as turning on a fan or air conditioner, removing external clothing, and reducing physical activity can all help to keep temperature constant.

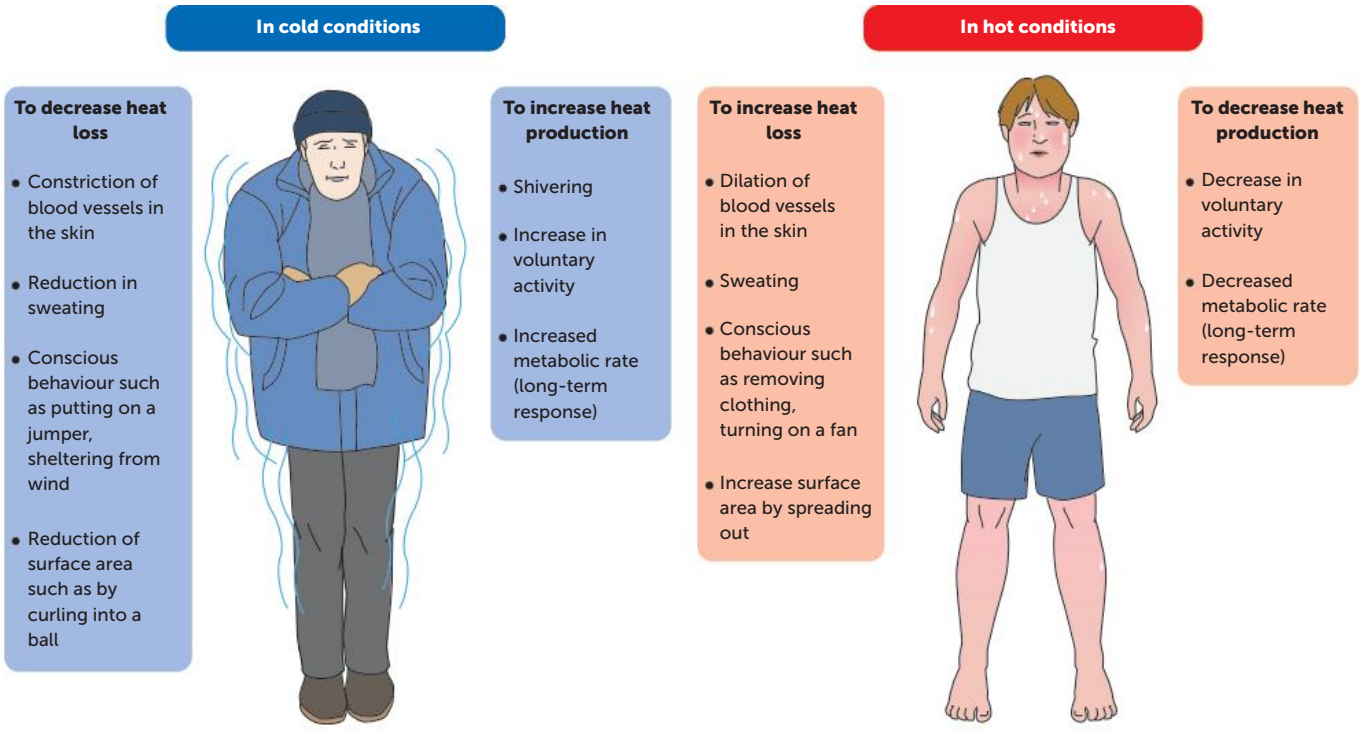

FIGURE 5.23 Mechanisms for regulating temperature

Control of thermoregulation

As we have seen, it is the hypothalamus that exercises control over the various mechanisms involved in maintaining body temperature. The hypothalamus monitors the temperature of the blood and receives impulses from the peripheral thermoreceptors. Through negative feedback loops involving the autonomic division of the nervous system, it controls the diameter of skin arterioles, sweating, shivering and other mechanisms for maintaining temperature. Figure 5.24 summarises the role of the hypothalamus in thermoregulation.

Activity 5.1
Investigating thermoregulation

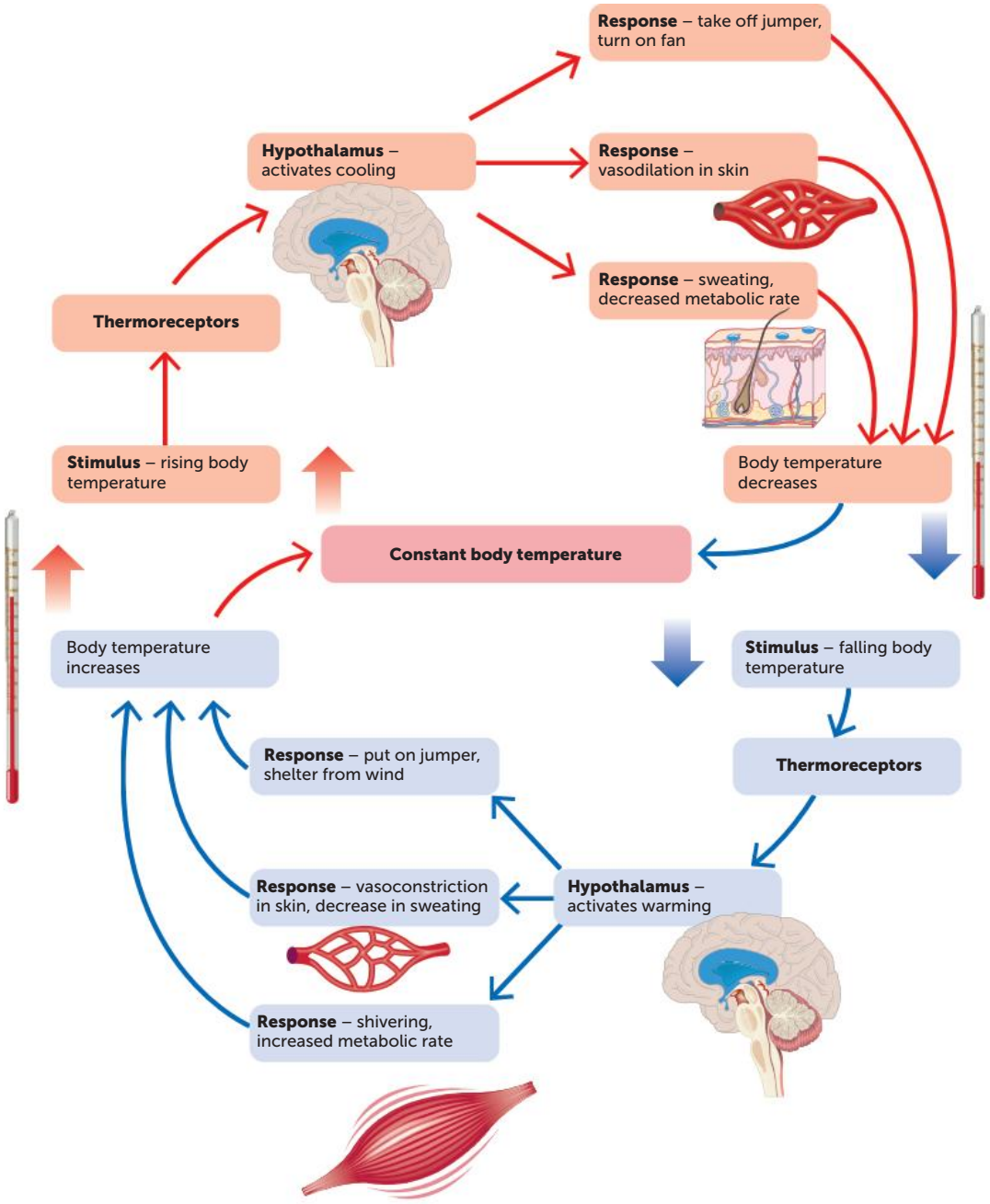

FIGURE 5.24
The role of the hypothalamus and mechanisms involved in maintaining a constant body temperature

Key concept

The hypothalamus receives information from thermoreceptors and initiates responses to maintain a core temperature of 36.8°C.

Temperature tolerance

A core body temperature over 42°C is dangerous and death usually occurs if it rises above about 45°C. High body temperature can result from a fever, but it can also occur in certain environmental conditions. When the temperature and relative humidity are high, it is difficult for the body to lose heat by radiation or evaporation. In this case, body temperature rises and the regulatory mechanisms cease. This is called **heat stroke** and it can be very serious, or even fatal, if brain cells are affected. Treatment consists of cooling the body as quickly as possible by immersing the patient in cold water.

Activity 5.2
Investigating experiments conducted in a heated room

Heat stroke
This website provides more information about heat stroke.

Heat exhaustion
This website provides more information about heat exhaustion.

Hypothermia
This website provides more information about hypothermia.

5.3 Homeostasis of body temperature

A condition that occurs more frequently than heat stroke is **heat exhaustion**. This condition occurs as a result of extreme sweating and vasodilation to lose heat. The loss of water in sweating reduces the volume of blood plasma and the vasodilation reduces resistance to blood flow. Blood pressure is thus reduced and output of blood from the heart decreases. The person may, therefore, collapse. Unlike in heat stroke, the body temperature is almost normal.

Extreme cold can also cause death. If a person's core temperature falls below 33°C, the metabolic rate is so low that heat production is unable to replace the heat lost and body temperature continues to fall. This condition is called **hypothermia**. Death can occur at core temperatures below 32°C, but people have been known to survive even lower temperatures.

Questions 5.3

RECALL KNOWLEDGE

1. Define 'thermoregulation' and state the set point for body temperature.
2. List the methods of heat loss.
3. Define 'metabolic rate' and list the reasons that the metabolic rate would:
   - **a** increase
   - **b** decrease.
4. State the location of thermoreceptors.
5. Why is the skin such an important organ in regulating body temperature?
6. Other than sweating, what other modes of evaporation occur in the body?
7. List five responses that the hypothalamus would trigger on a hot day. For each, classify them as responses from the nervous system or endocrine system.
8. Which is more dangerous – heat stroke or heat exhaustion? Explain why.

APPLY KNOWLEDGE

9. Heat will flow from areas of high temperature to low temperature until the temperatures are equal. Considering this process, explain how the body is able to maintain a temperature above that of the surrounding environment.
10. Use a Venn diagram to compare and contrast vasodilation and vasoconstriction.
11. Explain why sweating is more effective at cooling the body when the humidity is low.
12. Imagine that you forgot to take your sleeping bag on a camping trip in winter. Draw a flow chart to show the process that would occur as the temperature dropped overnight.
13. People who live in cold climates don't seem to 'feel' the cold as much as visitors to the area. Explain why this happens.

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CHAPTER 5 ACTIVITIES

ACTIVITY 5.1 Investigating thermoregulation

Working with a partner or in a small group, design an investigation to answer a question, or questions, about thermoregulation.

Some ideas include:

- making the subject as hot as possible by seating them in front of a heater, covering them with blankets and soaking their feet in buckets of warm water
- making the subject as cold as possible by removing any jumpers, wrapping their body in a wet towel and turning on a fan, soaking their feet in iced water and sitting them in the coolroom of the school canteen.

You should consider:

1. What characteristics can you measure to determine the effect of changing the temperature?
2. What changes occur in core body temperature when the skin temperature is changed?
3. What changes are evident at the surface of the body when the environment is very hot or very cold?
4. Do changes in environmental temperature affect breathing rate, heart rate or blood pressure?
5. What factors were you able to control?
6. What conclusion can you make from your investigation?

ACTIVITY 5.2 Investigating experiments conducted in a heated room

Sir Charles Blagden (1748–1820) was an English doctor and scientist. In a report to the Royal Society in 1775, he was the first person to describe the link between sweating and regulation of body temperature. He carried out many experiments on thermoregulation and in 1775 published his results in a paper titled 'Experiments and Observations in an Heated Room'. In one of his experiments he spent 45 minutes in a chamber that was heated to more than 120°C. With him in the chamber were some research assistants, a dog and a piece of beef. They all emerged from the chamber unharmed, but the beef was cooked.

1. Beef is mostly muscle tissue from a cow or a bull. Explain how the men's muscles were unharmed after 45 minutes in the hot chamber, while the muscle from the cow was cooked.
2. Would you expect to see any changes in the appearance of the men after they had been in the hot chamber? Explain your answer.
3. The men were in the hot chamber for 45 minutes. Do you think they would be able to survive for a much longer period? Explain.
4. Do you think the men (and the dog) would have had anything to drink while in the chamber? Explain.
5. A sauna is a small room where people can experience heating in dry or humid conditions. The temperature in the room can vary from 60°C to 120°C. High humidity is used at lower temperatures, but at higher temperatures only dry heat is used. Explain why a high-humidity sauna should not be set to a high temperature.
6. Suggest some precautions that should be taken when using a sauna.

Experiments and observations in a heated room

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CHAPTER 5 SUMMARY

- Homeostasis is maintenance of a constant internal environment, e.g. for temperature, blood glucose, pH and gas concentration. It is achieved by the endocrine system and the nervous system.
- A feedback system is a response that alters the original stimulus, providing feedback. In a negative feedback system, the response is the opposite of the original stimulus. In a positive feedback system, the response intensifies the original stimulus.
- Negative feedback systems (steady state control systems) maintain constant levels around the set point. The levels fluctuate between the tolerance levels and so the situation is called a dynamic equilibrium.
- A negative feedback system involves a stimulus, receptor, modulator, effector and feedback.
- Childbirth and blood clotting are examples of a positive feedback system.
- Glucose levels in the blood need to be between 4 and 6 mmol/L to supply the cells with glucose for cellular respiration.
- Insulin causes glucose to convert to glycogen by glycogenesis, which is stored in the liver and skeletal muscles.
- Under the influence of glucagon, glycogen can be broken down to produce glucose in the process of glycogenolysis.
- Glucose can also be produced from lipids and proteins in gluconeogenesis.
- The islets of Langerhans are hormone-producing cells in the pancreas. They contain alpha cells that secrete glucagon and beta cells that secrete insulin.
- Insulin is secreted when blood glucose levels rise. It lowers blood glucose levels by increasing the uptake of glucose by the cells, glycogenesis, and converting glucose to lipids and proteins.
- Glucagon is secreted in response to a decreased blood glucose level. It increases blood glucose levels by stimulating glycogenolysis, gluconeogenesis and protein breakdown.
- The adrenal cortex secretes glucocorticoids such as cortisol, which stimulates glycogenolysis and the breakdown of proteins and amino acids to increase blood glucose levels.
- The adrenal medulla secretes adrenaline and noradrenaline, which stimulate the conversion of glycogen to lactic acid and then glucose.
- Thermoregulation is the maintenance of body temperature at approximately 36.8°C by balancing heat loss and gain.
- Heat is produced during metabolism and absorbed from the surroundings. The rate of metabolism increases during exercise, with stress, and if body temperature increases.
- Heat is lost from the body by conduction, convection, radiation and evaporation.
- Thermoreceptors (either heat or cold receptors) detect changes in the temperature. Peripheral thermoreceptors detect the external temperature, while central thermoreceptors detect the internal temperature.
- The body loses heat via the skin. Blood in vessels in the dermis conduct heat to the outside of the body and sweat absorbs heat as it evaporates.
- To conserve heat, the hypothalamus triggers vasoconstriction; increased metabolism due to adrenaline, noradrenaline and thyroxine secretion; shivering and behavioural responses.
- To lose heat, the hypothalamus triggers vasodilation, sweating, decreased metabolism due to less thyroxine being secreted, and behavioural responses.
- Heat stroke occurs when the core body temperature is higher than 42°C.
- Heat exhaustion occurs when there is extreme vasodilation and sweating. The body temperature is almost normal, but low blood pressure may cause collapse.
- Hypothermia occurs when the body temperature falls below 33°C and the metabolic rate decreases.

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CHAPTER 5 GLOSSARY

Alpha cell A type of cell in the islets of Langerhans in the pancreas that secretes the hormone glucagon

Beta cell A type of cell in the islets Langerhans in of the pancreas that secretes the hormone insulin

Central thermoreceptor A thermoreceptor located in the hypothalamus

Cold receptor A receptor that is stimulated by low temperature

Conduction The transfer of heat by direct contact between particles

Convection The transfer of heat by the movement of liquids or gases

Cortex The outer part of an organ – for example, the adrenal gland

Cortisol A hormone that, along with related hormones, promotes normal metabolism

Dynamic equilibrium A state reached when the rates of forward and reverse changes are equal; a stable, balanced or unchanging system results

Evaporation The process of a liquid forming a gas, which absorbs heat energy

Feedback system A situation where the response to a stimulus changes the original stimulus

Glucagon A hormone secreted by the pancreas that increases blood sugar level

Gluconeogenesis The process of producing glucose molecules from lipids and amino acids

Glycogen A polysaccharide made up of thousands of glucose molecules bonded together in branching chains; functions as a store of glucose molecules in muscle and liver cells

Glycogenesis The process whereby glucose molecules are chemically combined in long chains to form glycogen molecules

Glycogenolysis The process of converting glycogen back to glucose

Heat exhaustion The collapse of a person after exposure to heat, during which their body's heat-regulating mechanisms continue to function normally

Heat receptor A receptor that is stimulated by high temperature

Heat stroke The failure of a person's temperature-regulating mechanisms when exposed to excessive heat

Homeostasis The maintenance of a relatively constant internal environment despite fluctuations in the external environment

Hypothermia Abnormally low body temperature; the temperature drops below the level required to maintain normal body functions

Insulin A hormone, secreted by the pancreas, that decreases blood sugar level

Islets of Langerhans Clusters of endocrine cells in the pancreas; secrete the hormones insulin and glucagon

Lipogenesis The production of lipids (fats)

Lipolysis The breakdown of lipids (fats) in the body

Medulla The inner part of an organ – for example, the adrenal gland

Metabolic rate The rate at which energy is released to the body by the breakdown of food

Model A simplified representation of an idea or a process; may be a diagram, flow chart, a simplified description of a complex situation or a physical model such as a model of a cell; examples are the stimulus–response–feedback model and the lock and key model for enzyme action

Modulator A control centre responsible for processing information received from a receptor and for sending information to the effector

Negative feedback Feedback that reduces the effect of, or eliminates, the original stimulus

Peripheral thermoreceptor A temperature receptor in the skin and in some mucous membranes

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Positive feedback Feedback that reinforces the original stimulus

Radiation The transfer of energy in the form of waves

Set point In a feedback system, the level at which a variable is to be maintained

Shivering Oscillating, rhythmic muscle tremors that increase body heat production

Steady state *see* homeostasis

Steady state control system A negative feedback system that maintains homeostasis

Stimulus Any change, internal or external, that causes a response

Sweat Fluid secreted by the sweat glands to help the body lose heat through evaporative cooling

Sweat gland A gland in the skin that produces sweat

Sweating The active secretion of fluid by the sweat glands

Thermoreceptor A temperature receptor; located in the skin or the hypothalamus

Thermoregulation The regulation of body temperature; the balance of heat gain and heat loss in order to maintain a constant internal body temperature independent of the environmental temperature

Tolerance limit The limit of factors such as temperature and fluid balance beyond which the body malfunctions

Vasoconstriction A decrease in the diameter of arterioles, restricting the flow of blood through them

Vasodilation An increase in the diameter of arterioles, increasing the flow of blood through them

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CHAPTER 5 REVIEW QUESTIONS

Recall

1 a What is homeostasis?
  b What aspects of the internal environment need to be regulated?
2 Define 'tolerance limits'.
3 Describe the role of the liver in regulating blood sugar concentration.
4 Distinguish between glycogenesis, glycogenolysis and gluconeogenesis.
5 Which gland is involved in the secretion of insulin and glucagon? Identify the location of the gland.
6 Describe the influence of the hormones of the adrenal glands on blood sugar concentration.
7 Describe the ways in which the body can gain heat.
8 What are the two types of thermoreceptors, and in what parts of the body are they located?
9 a What responses are likely to occur if core body temperature begins to fall?
  b What responses are likely to occur if core body temperature begins to rise?

Explain

10 Explain what the following terms mean and their relevance to homeostasis:
  a dynamic equilibrium
  b set point.
11 Why is the stimulus–response–feedback mechanism referred to as a model?
12 a Using examples, explain the difference between positive and negative feedback.
  b Why would a negative feedback loop be able to achieve homeostasis?
13 After a meal, the blood glucose level often rises well beyond the normal level. Explain why this occurs.
14 Explain how insulin and glucagon regulate the concentration of glucose in the blood.
15 Explain why heat loss must equal heat gain.
16 The skin plays an important role in thermoregulation. Explain how it is able to achieve this function.

Apply

17 Apply the stimulus–response–feedback model to the response of the pancreas after a chocolate bar is eaten.
18 Compile a table that summarises the role of each of the following systems in regulating blood glucose level: nervous system, digestive system, endocrine system, circulatory system, muscular system, excretory system.
19 Draw a stimulus–response model to show the processes involved with shivering.
20 In very cold weather, it is our fingers and toes that often feel coldest.
  a Why are fingers and toes affected by cold more than other parts of the body?
  b The fingers and toes may appear white when very cold. Explain why.
21 Alcohol increases blood flow through the skin. If a person suggested 'a stiff drink' would warm them up, what would you advise them, and why?
22 As a first aider at a sports carnival, it is important that you know the difference between heat stroke and heat exhaustion. Explain how you will distinguish between the two conditions.
23 A thermogram shows the temperature at the surface of an object or body. Examine the thermogram shown in Figure 5.17.
  a What parts of the ballerina's skin are the hottest? The coolest?
  b Explain the reasons for the differences in skin temperature that you have described in your answer to part a of this question.

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Extend

24 People with type 1 diabetes are unable to produce insulin.

- **a** Explain why patients have an increased blood glucose level.
- **b** One symptom of type 1 diabetes is tiredness. Explain why this occurs.

25 Vasoconstriction in the skin occurs when a person's body temperature is low, or when a person is very scared or very angry. (We say that someone is 'white with fear' or 'white with anger'.)

- **a** Apply the feedback model to each of these responses. Do they both fit the model? Explain your answer.
- **b** What is the advantage to a person of vasoconstriction in the skin:
  - **i** when their body temperature is tending to fall?
  - **ii** when they are scared or angry?

26 In the arms and legs there is exchange of heat between the arteries carrying blood to the limbs and the veins taking blood away from the limbs. This is called a countercurrent heat exchange. Find out how countercurrent heat exchange operates and describe its significance in maintaining core body temperature.

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6 HOMEOSTASIS CONTROLS FLUID AND GAS LEVELS

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including the collection of data related to homeostasis and the use of models of disease transmission, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

SCIENCE UNDERSTANDING

Homeostasis

- homeostatic processes involve nerves and hormones in maintaining the body's internal environment within tolerance limits through the control of metabolism and physiological and behavioural activities
- body fluid concentrations are maintained by balancing water and salts via the skin, digestive system and the kidneys, which involve the actions of antidiuretic hormone (ADH) and aldosterone on the nephron, and the thirst reflex
- gas concentrations are controlled by balancing the intake of oxygen and the removal of carbon dioxide via the lungs, through the actions of the medulla oblongata and the autonomic nervous system

Source: School Curriculum and Standards Authority, Government of Western Australia

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6.1 REGULATION OF THE COMPOSITION OF BODY FLUIDS

Water makes up a large proportion of the human body, ranging from 75% by mass in infants, to 50% for adult females and 60% for adult males, to 45% in old age. It is not pure water; rather, it contains dissolved substances. This fluid plays many important roles in the body, including transporting substances from one area of the body to another, facilitating movement across membranes and being the site of chemical reactions. Therefore, it is vital that the volume of water and concentration of dissolved substances is controlled to remain within the tolerance levels for the body.

Distribution of body fluids

The body of a male weighing 70 kg, with a water content of 60%, would contain 42 L of water. This large amount of water is distributed between the various body fluids.

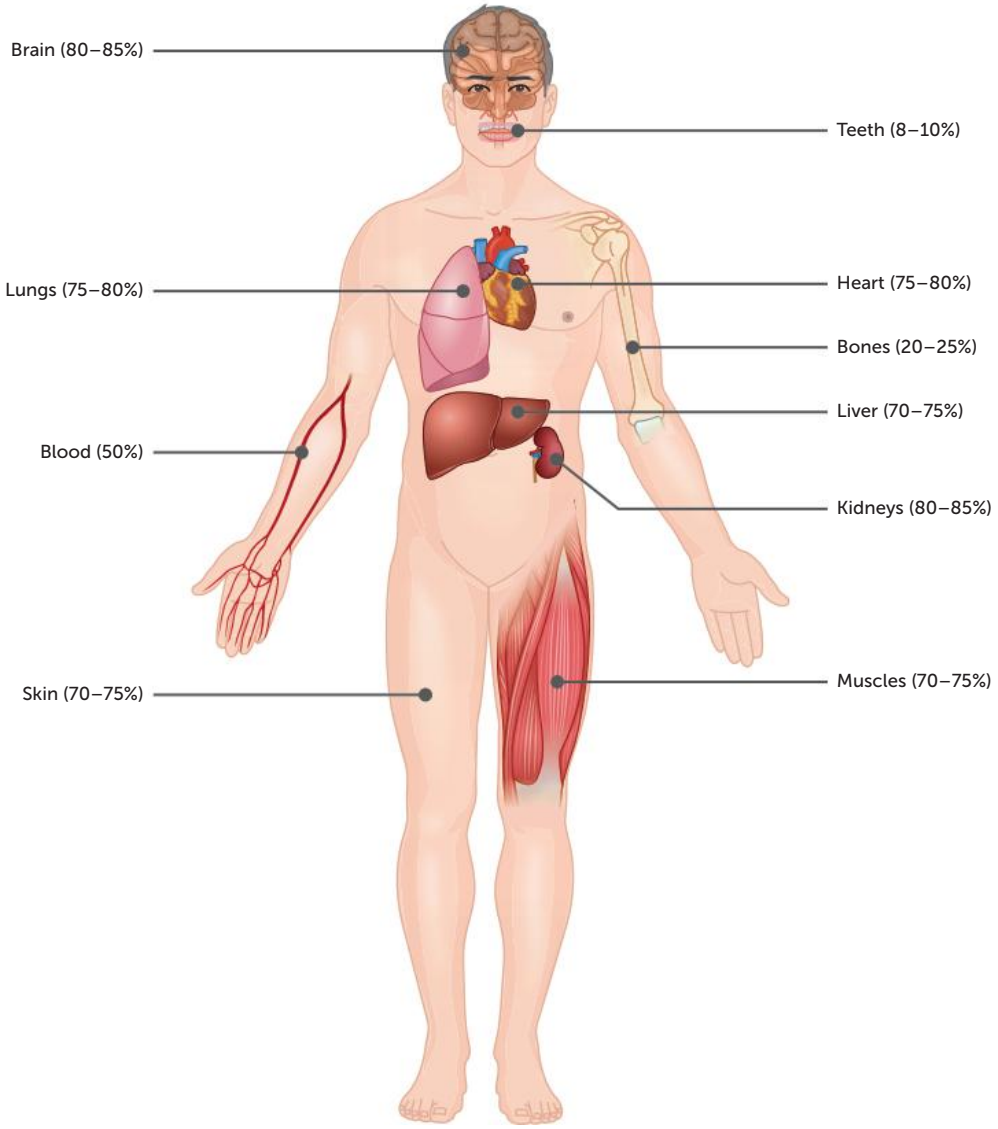

FIGURE 6.1 The water content of different body structures

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- Fluid inside the cells is called **intracellular fluid**, or **cytosol**.
- Fluid outside the cells is **extracellular fluid**. Extracellular fluid includes the:
  - blood plasma located within the blood vessels, also known as **intravascular fluid**
  - fluid between the cells, known as **interstitial fluid**, **intercellular fluid** or **tissue fluid**
  - fluid in specific body regions, known as **transcellular fluid**, which includes the fluid in the brain and spinal cord, eyes and joints, and surrounding the heart.

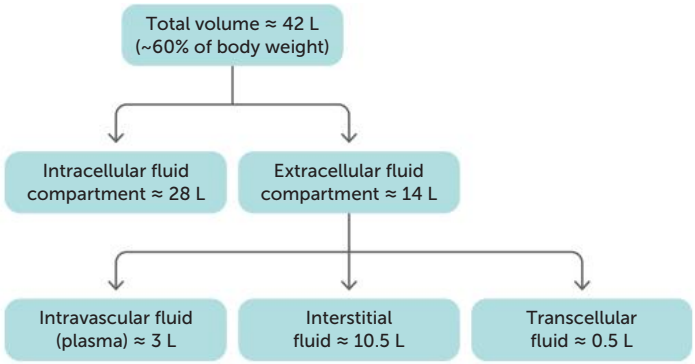

FIGURE 6.2 Distribution of body fluid in an average male adult

TABLE 6.1 Body fluids

TYPE OF BODY FLUID	PROPORTION OF TOTAL BODY FLUID	COMPONENTS OF THE BODY FLUID
Intracellular fluid	2/3 of total body water	Fluid inside the cell – the cytosol
Extracellular fluid	1/3 of total body water	Fluid that is outside the cells
Plasma (intravascular fluid)	Approx. 1/4 of extracellular fluid	The fluid part of the blood
Interstitial fluid and transcellular fluid	Approx. 3/4 of extracellular fluid	Lymph, cerebrospinal fluid, synovial fluid in joints, fluids of eyes and ears, fluid in the chest and abdominal cavities and around the heart, fluids of the alimentary canal, kidney filtrate

The different body fluids are not isolated from one another – there is a continuous exchange of materials between them. Plasma is separated from the interstitial fluid by the thin walls of the capillaries, and there is a relatively free exchange of materials between the two. However, dissolved materials that are large molecules, such as the proteins of the plasma, tend to remain within the blood vessels as they are too large to move through the capillary walls.

Water moves easily through plasma membranes, and so any difference in **osmotic concentration** between the intracellular fluid and the extracellular fluid does not last very long. If an imbalance in osmotic concentration does occur in any tissue, osmosis normally restores the balance within seconds. The tendency of a solution to take in water is known as **osmotic pressure**. The greater the difference in osmotic concentrations between two solutions, the greater the osmotic pressure. For more information about the exchange of materials across cell membranes refer to *Human Perspectives ATAR Units 1 & 2*.

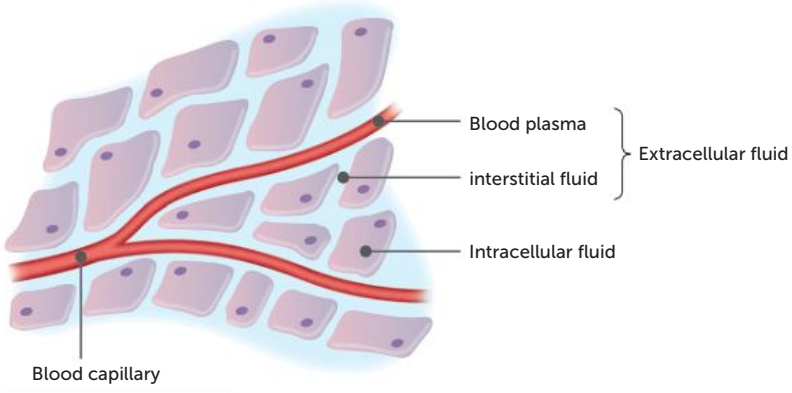

FIGURE 6.3 Body fluids

Osmoregulation and osmotic balance
This website has more information about osmotic pressure and osmosis.

Maintaining fluid balance

In the same way that heat gain must be equal to heat loss in order to maintain a constant body temperature, fluid gain must equal fluid loss if the composition of body fluids is to be kept fairly constant.

Most body fluid is obtained from water that is either taken in as a liquid or contained in food that is eaten. A small amount is obtained as a by-product of chemical processes occurring within the cells. This water is referred to as **metabolic water**.

Fluids are lost from the body via the kidneys, through the skin, from the surface of the lungs and from the alimentary canal. Typically, about 2.5 L of fluid are lost each day. Figure 6.4 summarises the sources of fluid intake and the avenues of output.

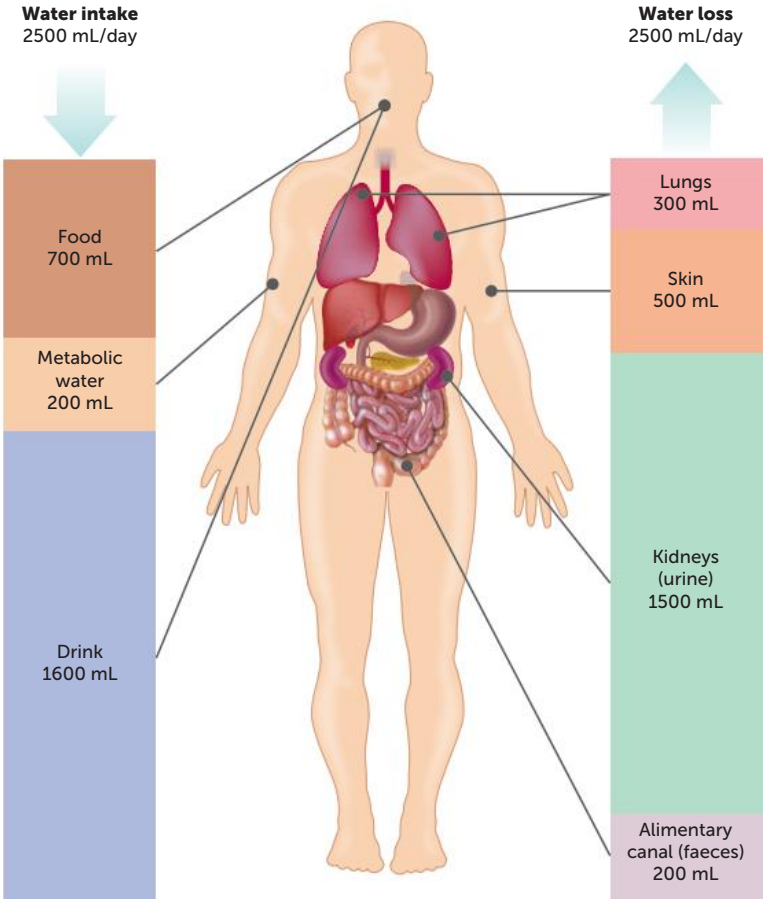

FIGURE 6.4 Daily fluid intake and output

Excretion

Excretion is the removal of the waste products of metabolism from the body. Many wastes are toxic and would be harmful to health if allowed to accumulate in the body fluids. Every cell produces waste products, so their removal before they reach harmful concentrations is extremely important.

Several organs in the body take part in excretion.

- The lungs are involved in the excretion of carbon dioxide. Carbon dioxide and water are produced by all body cells during cellular respiration. The body cannot use carbon dioxide and it is carried in the blood until it reaches the lungs, where it is excreted. Some water is also lost from the lungs, in the form of water vapour, as we exhale.
- Sweat glands in the skin secrete water containing by-products of metabolism such as salts, urea and lactic acid.

- The alimentary canal passes out bile pigments that entered the small intestine with the bile. These pigments are the breakdown products of haemoglobin from red blood cells. They leave the body with the faeces. The bulk of the faeces is composed of undigested food materials. These are not considered to be excretory products, as they have not been produced by the cells.
- The **kidneys** are the principal excretory organs. They are responsible for maintaining a constant concentration of materials in the body fluids. One of the important wastes removed by the kidneys is urea, which is produced in the liver during the breakdown of proteins.

Kidneys

As you can see from Figure 6.4, about 60% of the water lost from the body each day is excreted by the kidneys as urine. Water loss from the lungs and from the alimentary canal cannot be regulated. Water loss from the skin (sweat) is directly linked to temperature regulation. This means that only water loss from the kidneys can be regulated to achieve a constant concentration of dissolved substances in the body fluids. Thus, the kidneys are not just excretory organs; they also play a major role in regulating the composition of body fluids.

The kidneys are a pair of reddish-brown organs located in the abdomen. They are on either side of the vertebral column, at about the level of the lowest ribs, and are attached to the rear wall of the abdominal cavity. Each kidney is about 11 cm long and, due to the presence of the liver, the right kidney is usually slightly lower than the left. The kidneys are embedded in, and held in position by, a mass of fatty tissue.

A tube, the **ureter**, leaves each kidney and drains into a muscular reservoir, the **urinary bladder**, which empties to the outside through another tube, the **urethra**.

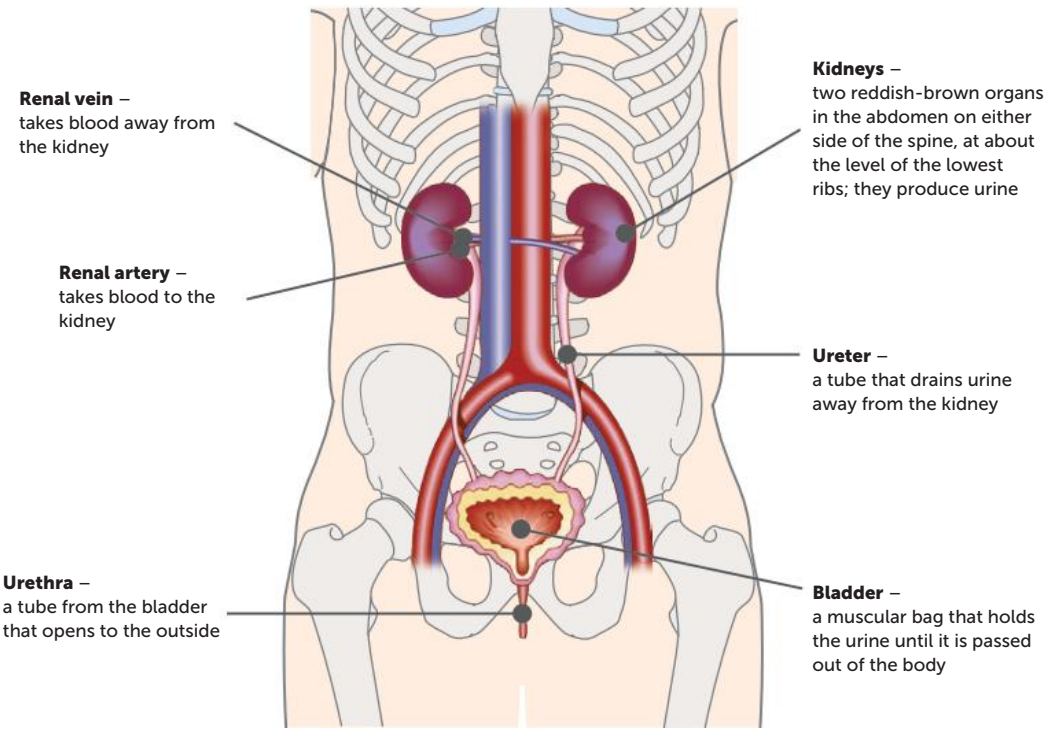

FIGURE 6.5 Kidneys and associated organs

Each kidney contains about 1.2 million microscopic units called nephrons. The **nephron** is the functional unit of the kidney; that is, it is the nephrons that carry out the kidney's role in excretion and water regulation. Figure 6.6 shows a nephron and explains how it functions. Detailed information about the structure and function of the nephron was covered in *Human Perspectives ATAR Units 1 & 2*.

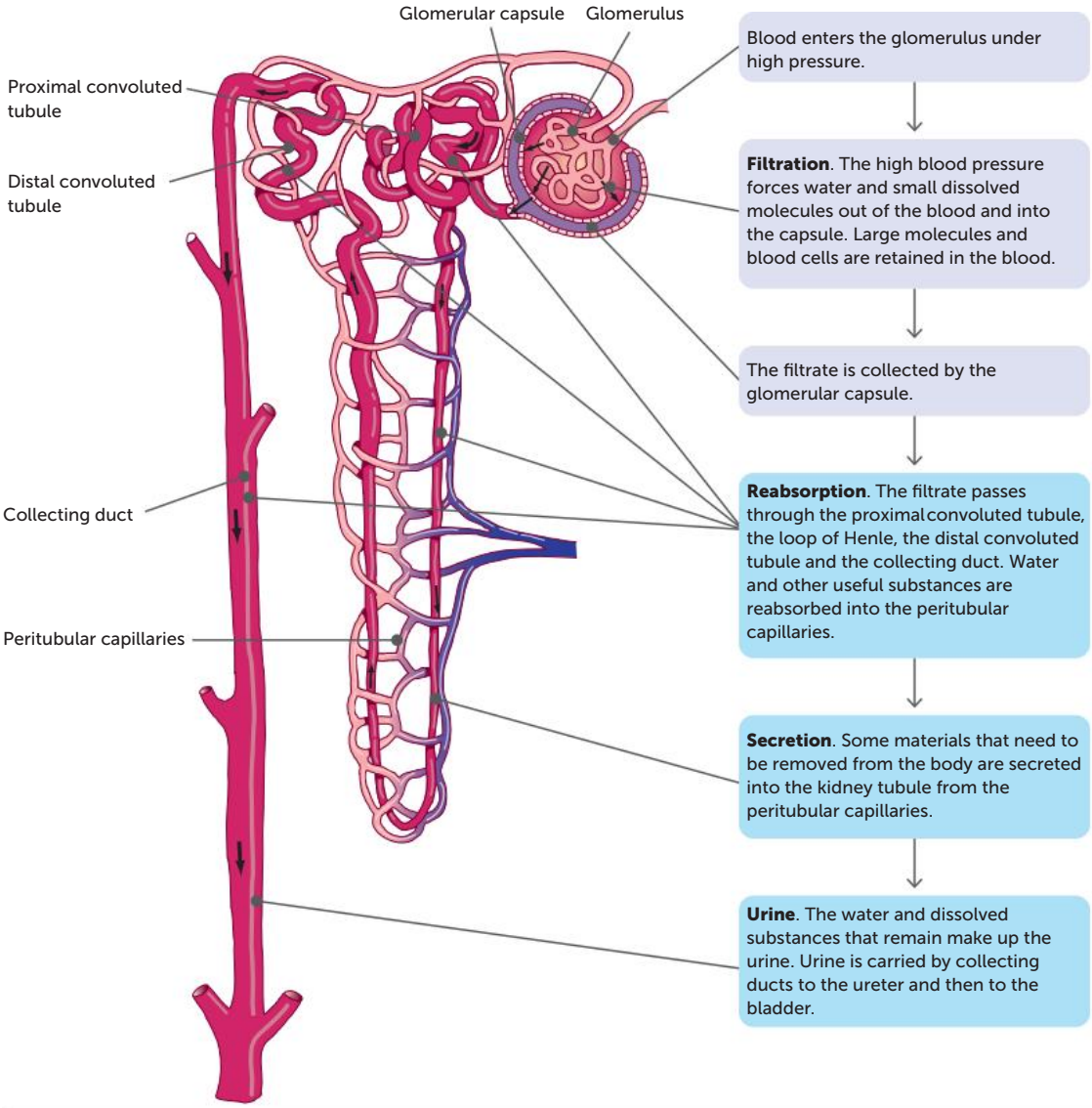

FIGURE 6.6 The functional unit of the kidney is the nephron

Key concept

The nephrons in the kidneys play a vital role in controlling the water content of the body.

Controlling water levels

Water is continually lost from the body in sweat, urine, faeces and exhaled breath. Normally, this is balanced by water intake. However, at times of strenuous activity or extreme heat this water loss can be quite high. As water is lost, the plasma becomes more concentrated and hence has a higher osmotic pressure. As a result, water moves from the interstitial fluid into the plasma by osmosis. This makes the interstitial fluid more concentrated and, therefore, water diffuses out of the cells, so the cells start to shrink from dehydration. When this happens, **osmoreceptors** in the hypothalamus detect the increase in osmotic pressure. A number of responses are then triggered that increase the water content and, hence, lower the osmotic pressure.

The kidneys and antidiuretic hormone

The volume and composition of urine produced by the kidneys depends on how much water there is in the body fluids. If you drink a large volume of water, you will quite soon produce a large volume of dilute urine. If you become dehydrated by not drinking enough water, you will produce a smaller volume of concentrated urine.

Approximately 99% of the water filtered through the glomeruli of the kidneys is reabsorbed. This reabsorption occurs through the walls of the kidney tubules along their entire length. The reabsorption occurring at the proximal convoluted tubule and loop of Henle is by osmosis, while reabsorption at the distal convoluted tubule and collecting tubule is active reabsorption. The level of active reabsorption is controlled by a hormone known as **antidiuretic hormone (ADH)**.

ADH is produced by the hypothalamus and released from the posterior lobe of the pituitary. The permeability of the walls of the distal convoluted tubule and collecting duct is controlled by ADH. When the concentration of ADH in the blood plasma is high, the tubules are very permeable to water, and thus water is able to leave the tubule and enter the surrounding capillary network. This outward flow of water from the fluid within the tubules reduces its volume and hence increases the concentration of the materials remaining. On the other hand, when the concentration of ADH in the plasma is low, the tubules are not very permeable to water, and little water is reabsorbed into the plasma of the blood. In this situation, the fluid within the tubules remains fairly dilute, as its volume is not reduced to any significant extent.

The action of ADH in controlling water balance is another example of a feedback process maintaining the internal environment of the body. For example, if water was lost through excess sweating, the following process would occur.

- *Stimulus:* The osmotic pressure of the blood is raised due to the decrease in water in the blood.
- *Receptors:* Osmoreceptors in the hypothalamus detect the increased osmotic pressure of the blood.
- *Modulator:* The hypothalamus stimulates the posterior lobe of the pituitary gland to release ADH into the bloodstream.
- *Effector:* ADH is carried all over the body by the blood but it affects its target organs, which are the nephron tubules in the kidney. The permeability to water of the distal convoluted tubules and the collecting ducts is increased.
- *Response*: More water is then reabsorbed into the blood plasma from the distal convoluting tubule and collecting duct.
- *Feedback:* The reabsorption of water increases the amount of water in the plasma and so the osmotic pressure of the blood is decreased. This adjustment has eliminated or reduced the original stimulus; a negative feedback has occurred.

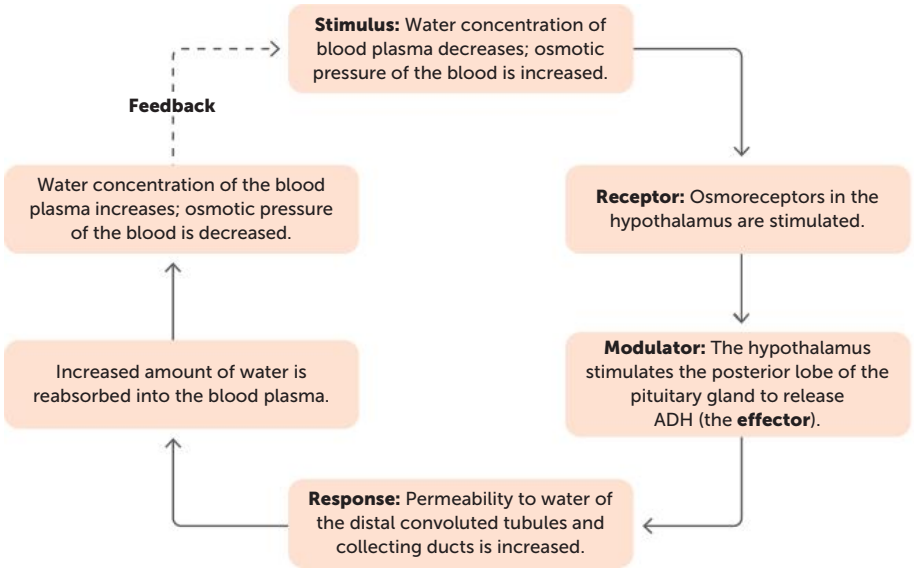

FIGURE 6.7 Regulation of water output by antidiuretic hormone

The kidneys and aldosterone

Aldosterone is another hormone that plays a part in the regulation of water output. Aldosterone, sometimes called the salt-retaining hormone, is secreted by the adrenal cortex in response to a:

- decrease in the concentration of sodium ions in the blood
- decrease in blood volume
- decrease in blood pressure
- increase in the concentration of potassium ions in the blood.

Aldosterone acts on the distal convoluted tubules and collecting ducts to increase the amount of sodium ions reabsorbed into the bloodstream and the amount of potassium secreted in the urine. It achieves this through active transport using a sodium-potassium pump. For every three sodium ions reabsorbed, two potassium ions are secreted. There is thus a net movement of ions into the blood and the subsequent transport of water into the blood via osmosis. Therefore, aldosterone has a role in regulating water content of the body.

The thirst response

In addition to reducing water loss, the water level of the body can be increased by taking in more fluid. Osmoreceptors are able to stimulate the **thirst centre** in the hypothalamus, prompting the person to drink water. This fluid is absorbed across the wall of the alimentary canal into the blood, decreasing the osmotic pressure.

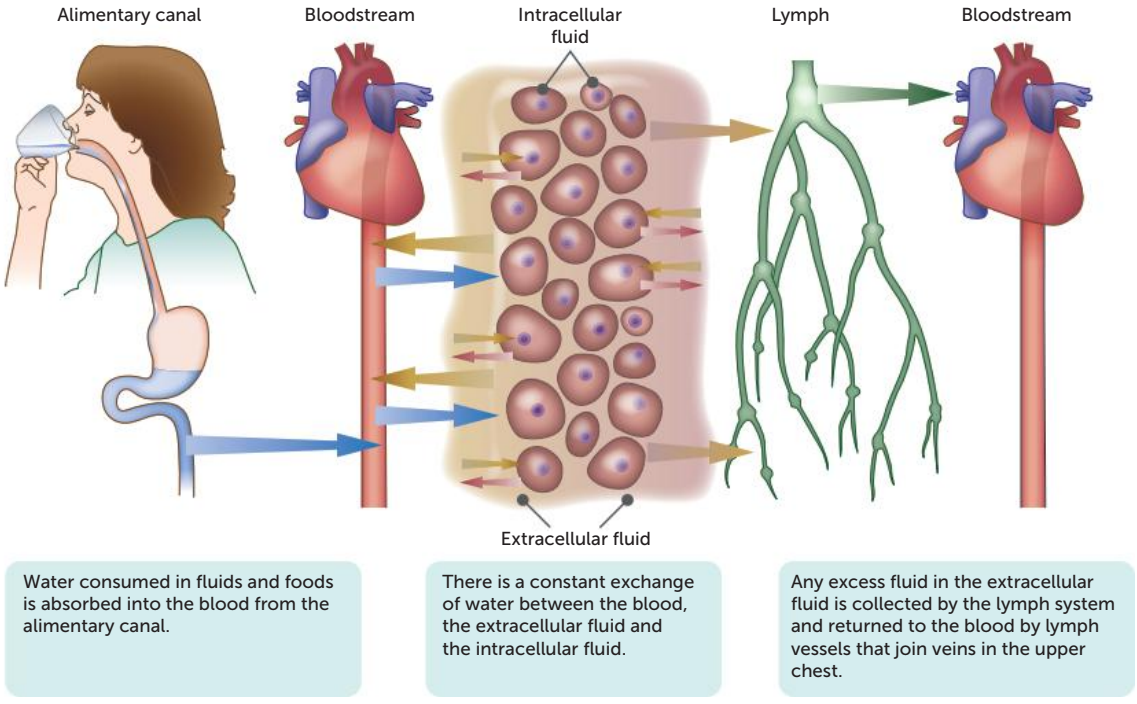

FIGURE 6.8 Movement of water between the various parts of the body

- *Stimulus:* As water is lost from the various body fluids, there is a reduction in plasma volume and an increase in osmotic concentration of the extracellular fluid.
- *Receptor:* Osmoreceptors in the thirst centre in the hypothalamus detect the rising osmotic concentration of the blood. Other stimuli such as a dry mouth are also involved.
- *Modulator:* Stimulation of the thirst centre in the hypothalamus makes the person feel thirsty.
- *Effector:* The conscious feeling of thirst stimulates the person to drink.
- *Response:* The fluid consumed is absorbed from the alimentary canal into the plasma in the blood.
- *Feedback:* As the blood circulates through the body, it enables the interstitial fluid and intracellular fluid to return to the normal osmotic concentration. After drinking, the thirst centre is no longer stimulated and the desire to take in water ceases.

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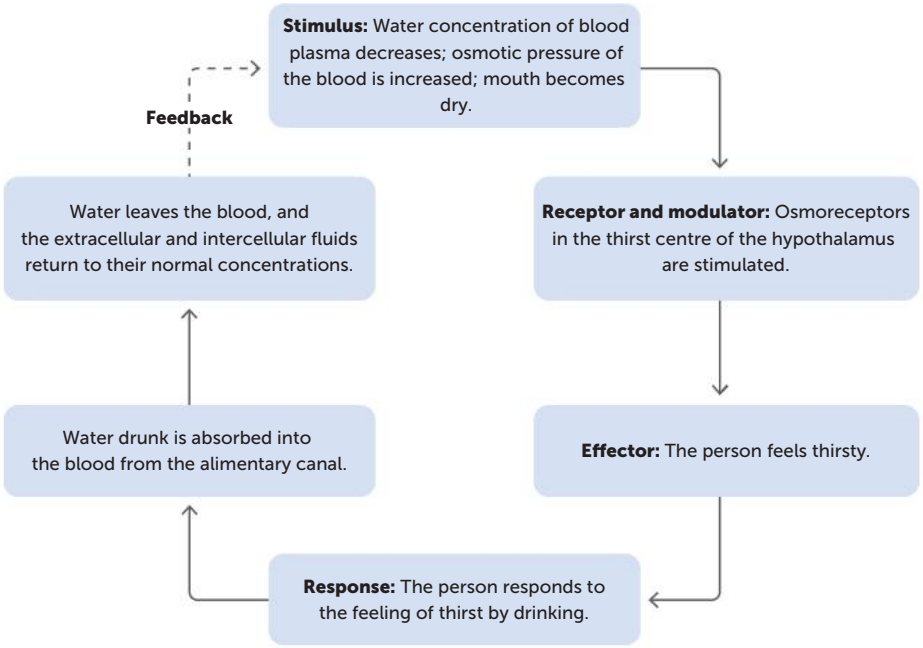

FIGURE 6.9
Regulation of water balance by the thirst mechanism

Activity 6.1
Simulated urinalysis

Key concept

The water content of the body is altered by the action of antidiuretic hormone and aldosterone on the kidneys, as well as by the thirst centre in the hypothalamus.

Too much and too little water

You have probably read stories or news reports about people dying in the desert through lack of water. When water loss exceeds water intake, there is not enough water in the body for it to carry out normal functions. This is called **dehydration**. Symptoms of dehydration become noticeable when a person has lost about 2% of their normal body water. The loss may be through sweating, vomiting or diarrhoea. Elderly people can suffer from dehydration because the thirst reflex becomes less effective as we grow older. Symptoms of dehydration include severe thirst, low blood pressure, dizziness and headache. If the condition remains untreated the patient becomes delirious, loses consciousness and dies.

It is also possible to have *too much* water in the body. This is called **water intoxication**, or sometimes water poisoning. It occurs when body fluids become diluted and cells take in extra water by osmosis. This may happen if a person loses a lot of water and salts through sweating and replaces the loss with plain water. In such cases, the water consumed should contain dissolved substances to replace the lost salts as well as the water. The first sign of water intoxication is usually lightheadedness. Headache, vomiting and collapse may follow.

Water intoxication
This website provides more information about water intoxication.

Strange but true: Drinking too much water can kill
An article about drinking too much water.

Dehydration
This website provides more information about dehydration.

What you should know about dehydration
Another article about dehydration.

6.1 Homeostasis of body fluids

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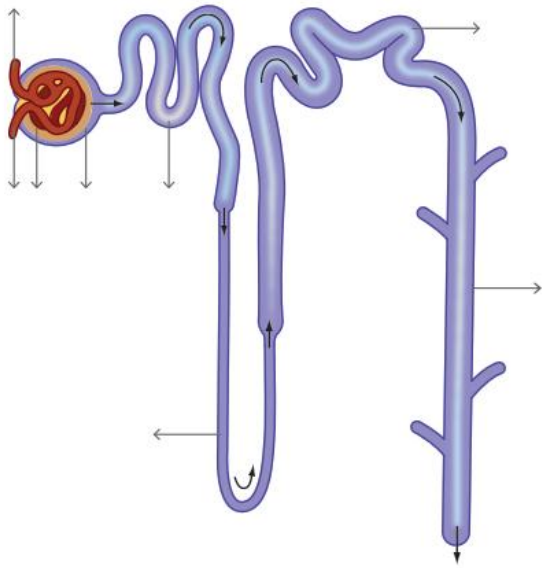
Questions 6.1

RECALL KNOWLEDGE

1. List three functions of water in the body.
2. What organ has the greatest percentage of water?
3. Complete the following sentence.

   The _____ is the fluid found outside the cells. It is made up of the intravascular fluid, which is also known as _____, the _____ fluid between the cells and transcellular fluid – for example, _____ (CSF).
4. True or false? There is more fluid found in the plasma than in the cells.
5. Describe osmosis.
6. List the ways that fluid is:
   - **a** gained
   - **b** lost.
7. Label the parts of a nephron on the diagram below.



8. Name the receptors that detect a change in water content of the body, and state where they are located.
9. **a** What does 'ADH' stand for?
   - **b** What part of the nephrons does ADH act on?
   - **c** What effect does ADH have on the structures identified in **b**?
   - **d** Is ADH secreted due to a high or low osmotic pressure?
10. Describe the role of aldosterone in maintaining the concentration of sodium ions in the blood. Identify any relevant structures in your description.

APPLY KNOWLEDGE

11. What is the mass of water in an adult female weighing 62 kg?
12. Suggest why the proportion of water in a baby's body is larger than in their grandparent's body.
13. Explain why water toxicity can cause cells to swell.
14. Explain why the homeostatic mechanisms that control body fluids are focused on the kidneys and not the alimentary canal, lungs or skin, even though water is also lost through these organs.
15. Draw a feedback loop to show what would happen if you drank more water than you lost.
16. Explain why, after running a cross-country course, Alex had a dry throat and was very thirsty.

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6.2 REGULATION OF GAS CONCENTRATIONS

Cellular respiration occurs in cells to provide energy for its functions. As it uses oxygen and produces carbon dioxide, cells need a continuous supply of oxygen and removal of carbon dioxide. Therefore, it is crucial that the levels of these gases in the body are regulated.

$$\text{Oxygen + glucose} \rightarrow \text{carbon dioxide + water + energy}$$
$$6O_2 + C_6H_{12}O_6 \rightarrow 6CO_2 + 6H_2O + \text{energy}$$

The respiratory system is responsible for taking in oxygen and excreting carbon dioxide from the body. In particular, the lungs are the organs in which the exchange of carbon dioxide for oxygen occurs. Therefore, changes in breathing change the amount of oxygen taken in and the amount of carbon dioxide excreted.

The circulatory system carries oxygen from the lungs to the cells, where it is used. It also takes away the carbon dioxide produced and delivers it to the lungs for excretion from the body. Thus, the circulatory system is also involved in the regulation of gas concentrations.

Control of breathing

The muscles that cause air to move in and out of the lungs are:

- the diaphragm, a muscle that separates the thorax from the abdomen
- the intercostal muscles, the muscles between the ribs.

These are skeletal muscles and require stimulation from nerve impulses to initiate contraction. The diaphragm is stimulated by impulses from the phrenic nerve, while impulses from the intercostal nerves stimulate the intercostal muscles. These spinal nerves have their origin in the spinal cord at the level of the neck and thorax.

The nerve impulses that travel to the diaphragm and intercostal muscles are controlled by a **respiratory centre** located in the medulla oblongata of the brain. There are two regions within the respiratory centre: one that controls expiration (breathing out) and one that controls inspiration (breathing in). To coordinate breathing, messages need to pass back and forth between the neurons in these two regions.

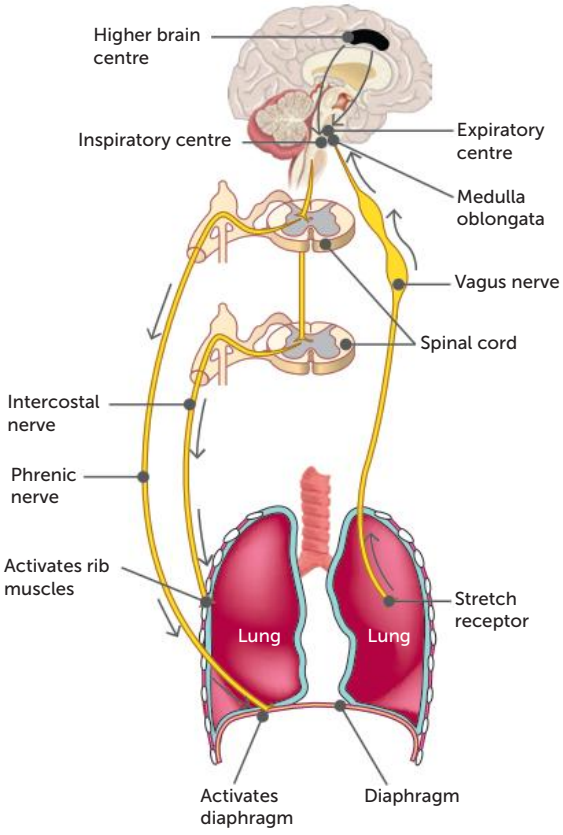

FIGURE 6.10 Control of breathing by respiratory centres in the brain

Chemicals affecting breathing

Both oxygen and carbon dioxide are carried in the blood and their concentrations affect the breathing rate and depth. In addition, the concentration of carbon dioxide in the blood plasma affects the concentration of hydrogen ions (H^+). When carbon dioxide dissolves in water, it forms carbonic acid (H_2CO_3), which breaks down readily to form hydrogen ions and bicarbonate ions (HCO_3^-), as shown in the following chemical equation.

$$CO_2 + H_2O \rightleftharpoons H_2CO_3 \rightleftharpoons H^+ + HCO_3^-$$

Oxygen, carbon dioxide and hydrogen ions all have some effect on the regulation of breathing activity.

Chemoreceptors

There are two types of **chemoreceptors**: peripheral chemoreceptors and central chemoreceptors.

- *Peripheral chemoreceptors* are groups of cells within the walls of the aorta and carotid arteries that are sensitive to changes in the concentration of oxygen, carbon dioxide and hydrogen ions in the blood plasma. These are known as the **aortic** and **carotid bodies**.
- *Central chemoreceptors* are located in the medulla oblongata. These are sensitive to changes in the concentration of carbon dioxide in the blood and hydrogen ions in the cerebrospinal fluid.

When chemoreceptors are stimulated, they send a nerve impulse to the area of the respiratory centre that regulates breathing.

Oxygen concentration

As oxygen is consumed by the cells, its concentration in the blood begins to fall. If the concentration of oxygen falls below normal while other factors are held constant, the breathing rate increases. However, within the normal range of blood oxygen concentration, the effect on breathing rate is only slight. The concentration has to fall to very low levels before it has a major stimulatory effect. Thus, under normal circumstances, oxygen plays little part in the regulation of breathing.

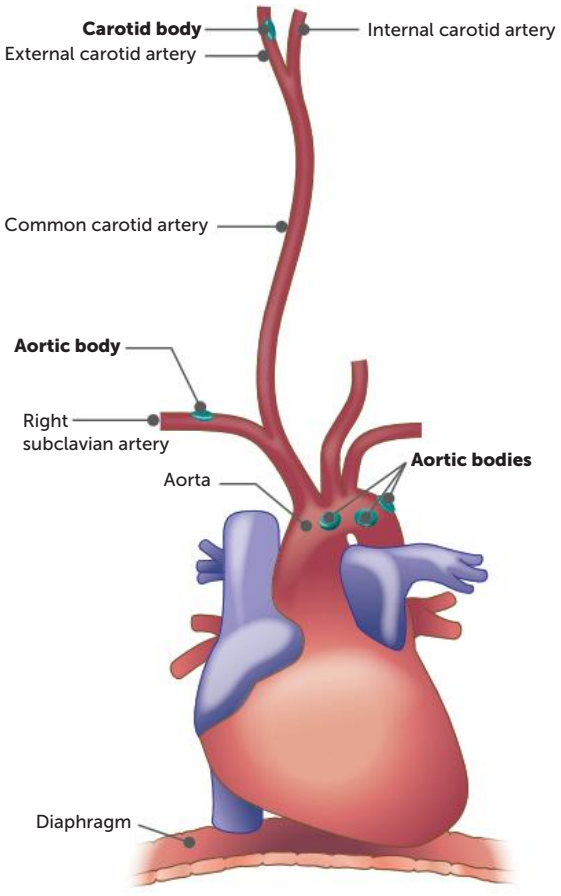

FIGURE 6.11 Location of the aortic and carotid bodies

A large decrease in oxygen concentration stimulates the peripheral chemoreceptors, and nerve impulses are transmitted to the respiratory centre. These nerve impulses stimulate the transmission of messages to the diaphragm and intercostal muscles, and so the breathing rate and depth increases.

Carbon dioxide concentration

The concentration of carbon dioxide in the blood plasma is a major factor in the regulation of breathing rate. A relatively small increase in the concentration of carbon dioxide is enough to cause a marked increase in the rate and depth of breathing.

As mentioned above, the concentration of carbon dioxide in the plasma is associated with the concentration of hydrogen ions. Any increase in carbon dioxide results in an associated increase in hydrogen ion concentration. The increase in concentration of both these chemicals in the blood stimulates the central and peripheral chemoreceptors. These in turn transmit nerve impulses to the respiratory centre, resulting in an increase in breathing rate and depth.

The chemoreceptors most sensitive to changes in the concentration of carbon dioxide in the plasma are those located in the medulla oblongata. These chemoreceptors are responsible for 70–80% of the increase in breathing rate that results from an increase in the carbon dioxide concentration of the blood. However, this response takes several minutes.

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The immediate increase in breathing rate that occurs following an increase in the carbon dioxide concentration of the plasma is produced by the stimulation of the aortic and carotid bodies. These bodies are stimulated by the associated increase in hydrogen ion concentration, as described below.

Hydrogen ion concentration

As the hydrogen ion concentration of the blood increases, the pH decreases. A decrease in the pH directly stimulates chemoreceptors in the aortic and carotid bodies, which then transmit impulses to the respiratory centre, resulting in an increase in the breathing rate and depth.

The regulation of the breathing rate in response to changes in the concentration of carbon dioxide and hydrogen ions (pH) is illustrated in Figure 6.12.

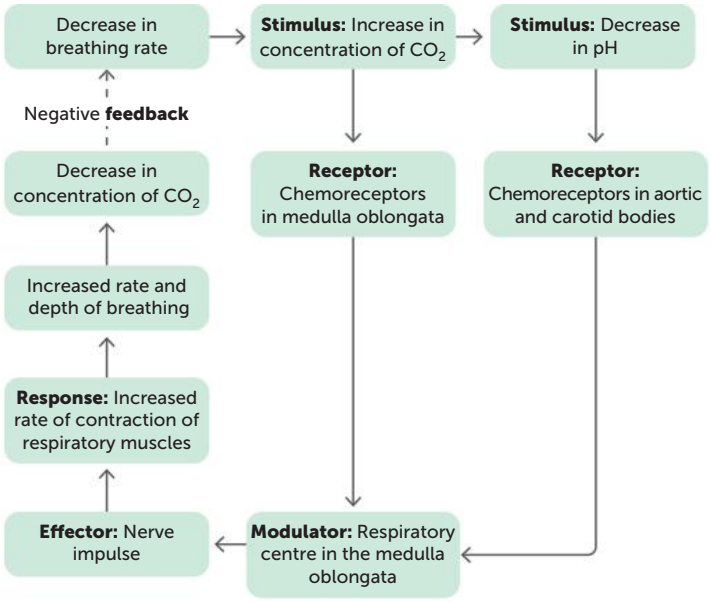

FIGURE 6.12
Negative feedback control of breathing rate through changes in the concentration of carbon dioxide and the pH of the blood plasma

None of the three factors (oxygen concentration, carbon dioxide concentration and hydrogen ion concentration) is independent in the regulation of breathing rate. Each factor interacts with the others. Nor are these the only factors to play a role in the control of breathing. At any instant, therefore, the rate of breathing is regulated by a number of factors, and the sensitivity of some factors, such as oxygen, is generally not as great as the sensitivity of others, such as carbon dioxide.

6.2 Homeostasis of gas concentrations

Key concept

Chemoreceptors recognise changes in the concentration of oxygen, carbon dioxide and hydrogen ions in the blood. They send nerve impulses to the respiratory centre in the medulla oblongata, which controls the rate and depth of breathing. Messages are sent from the respiratory centre, along the descending tract of the spinal cord and then to the phrenic and intercostal nerves to cause the contraction and relaxation of the diaphragm and intercostal muscles. Increasing the rate of the cycle of contraction and relaxation will increase the rate and depth of breathing and, therefore, the inhalation of oxygen and exhalation of carbon dioxide. This, in turn, will increase the concentration of oxygen and decrease the concentration of carbon dioxide.

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Voluntary control of breathing

Humans are able to voluntarily control their rate and depth of breathing, factors very important in speech. We can even stop breathing for a limited period. This voluntary control comes via connections from the cerebral cortex to descending tracts in the spinal cord. Voluntary control thus bypasses the respiratory centre in the medulla oblongata. This is a protective device as it enables us to prevent irritating gases and water from entering the lungs.

Holding our breath

While it is possible to hold our breath, we cannot stop breathing forever! The build-up of carbon dioxide in the plasma stimulates the inspiratory centre to send impulses to the inspiratory muscles. Thus, we are eventually forced to take a breath whether we want to or not.

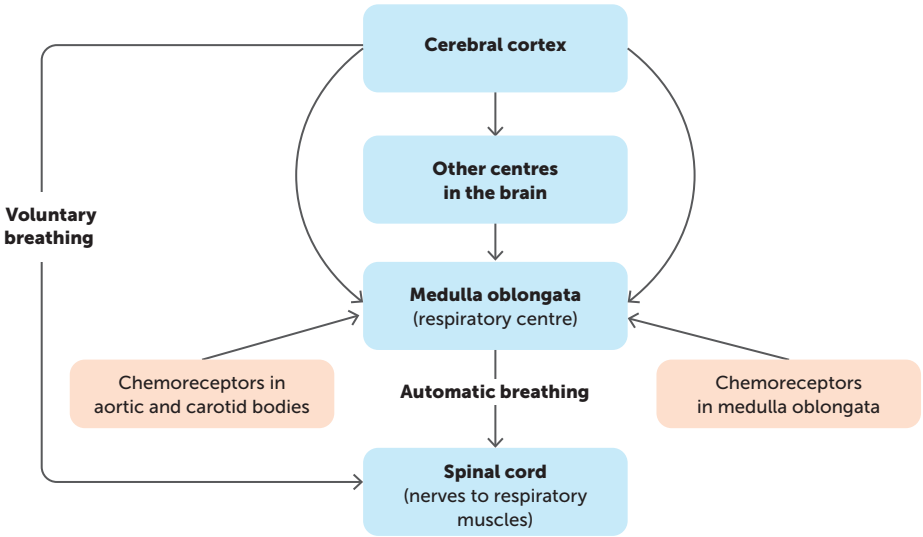

FIGURE 6.13 Voluntary and involuntary control of breathing by the central nervous system

Hyperventilation

Rapid, deep breathing can provide more oxygen than required and remove more carbon dioxide than necessary. This is called **hyperventilation**. It can occur voluntarily or may be stimulated by physical stress such as severe pain or emotional stress such as extreme anxiety. Hyperventilation usually corrects itself because the reduction in carbon dioxide concentration means that the chemoreceptors are not stimulated, reducing the rate and depth of breathing until carbon dioxide levels return to normal.

A very dangerous practice is to hyperventilate before swimming under water. Although this practice does allow a person to stay under water longer, it is not because of extra oxygen in the blood; it is due to the loss of carbon dioxide. The breath-holding ability could be increased to such an extent that the individual loses consciousness from lack of oxygen to the brain before feeling the urge to breathe. Many drowning deaths in Australia have been the direct result of hyperventilation.

Respiration control
This website provides more information on the regulation of breathing.

Activity 6.2
Investigating breathing rate

Activity 6.3
Investigating behaviour and homeostatic mechanisms

Exercise and breathing

During exercise, the contracting muscle cells require large amounts of oxygen and produce large amounts of carbon dioxide. In responding to this increased demand for gas exchange, the respiratory system increases both the rate and depth of breathing.

During heavy exercise, the volume of air going into and out of the lungs each minute may increase 10- to 20-fold. The same factors that influence breathing at rest appear to be involved in this increase – that is, the concentrations in the blood plasma of carbon dioxide, hydrogen ions and, to a lesser extent, oxygen.

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Questions 6.2

RECALL KNOWLEDGE

1 What is the name of the peripheral chemoreceptors that influence breathing?
2 Write the word equation for cellular respiration.
3 Describe the relationship between carbon dioxide and the concentration of hydrogen ions in the blood.
4 Which nerve sends impulses to:
   a the diaphragm?
   b the intercostal muscles?
5 Which chemoreceptors detect changes in:
   a oxygen?
   b carbon dioxide?
   c hydrogen ions?
6 Explain how an increased rate and depth of breathing is able to decrease the concentration of carbon dioxide in the blood.
7 Explain why it is not possible to die from holding your breath unless you are under water.

APPLY KNOWLEDGE

8 Use a flow chart to show the pathway of messages from the respiratory centre to the diaphragm during inspiration.
9 Why is it important that the muscles responsible for breathing are skeletal muscles, and not smooth or cardiac muscles?
10 Draw a feedback loop to show what would happen in the body if the concentration of oxygen decreased by a:
   a small amount
   b large amount.
11 Both central and peripheral chemoreceptors respond when there is an increase in carbon dioxide levels. Explain why it is important that both of these receptors are stimulated.
12 People with chronic obstructive pulmonary disease have inflamed airways that make it difficult to breathe. This can lead to a condition known as hypercapnia, an increased level of carbon dioxide.
   a One symptom of hypercapnia is that the blood becomes acidic. Explain why this would occur.
   b Explain why giving oxygen to patients with hypercapnia could be dangerous.

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CHAPTER 6 ACTIVITIES

Developed exclusively by Southern Biological

ACTIVITY 6.1 Simulated urinalysis

In this investigation, you are tasked with performing a urinalysis. You will test the colour, pH, glucose, protein and specific gravity of three known urine samples. The known urine samples are low, normal and high simulated urine samples that are engineered to produce results indicative of those characteristics. Following this, you are also tasked with conducting a urinalysis of two unknown simulated urine samples. You will be required to interpret the results and identify whether the results indicate any diseases.

Aim

To perform a simulated urinalysis and determine the characteristics of two unknown samples
Time requirement: 30 minutes

You will need

Simulated urine samples 100 mL approx. (low, normal, high, unknown A and unknown B); graduated cylinder; 5 plastic urine specimen containers; marker; 5 pH test strips and pH colour chart; 5 urine reagent strips; hydrometer and jar; disposable gloves; paper towel

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Simulated urine can cause skin or eye irritation.	Avoid any direct contact. If contact does occur, immediately flush the affected area with water.
Simulated urine can stain clothing and skin.	Wear gloves at all times and avoid any direct contact.

What to do

Colour and pH testing

1. Collect three plastic urine specimen containers. Using a marker, label them Low, Normal and High.
2. Using a graduated cylinder, transfer 10 mL of Low, Normal and High simulated urine into the corresponding urine specimen containers. Wash the measuring cylinder between each measurement if using the same equipment for each.
3. Inspect the samples and note the colour of each sample in Table 1.
4. Collect three pH test strips and label one end of each 'L', 'N' and 'H', respectively.
5. Dip the 'L' strip into the 'Low' urine sample. Wipe off any excess liquid against the side of the container and lay the strip on a paper towel. Compare the colour of the test strip to the pH colour chart and record the results in Table 1.
6. Repeat step 5 for the 'Normal' and 'High' urine samples using the corresponding pH test strip and record the results in Table 1.

Glucose and protein testing

1. Collect three reagent strips and label the end of each strip that does not contain a test square. Label them 'L', 'N' and 'H', respectively.
2. Observe the colour of the test squares that are attached at one end of the urine reagent strip and determine which square tests for glucose and which tests for protein.

3 Dip the test square end of the 'L' strip into the 'Low' urine sample and then withdraw it. Wipe off any excess liquid against the side of the container and lay the strip on a paper towel. After 30 seconds, a colour change on either square will indicate a positive result for that test, and no colour change will indicate a negative result. Refer to the product information/colour chart provided with the testing strips. Record the results in Table 1.
4 Repeat step 3 for the 'Normal' and 'High' urine samples using the corresponding test strip and record the results in Table 1.

Specific gravity testing

1 Clean the urine hydrometer and jar thoroughly.
2 Fill the jar to the fill line (as marked by the teacher) with the 'Low' urine sample and insert the hydrometer into the jar.
3 Note the fluid level on the hydrometer scale. Add or subtract the calibration factor to your measurement (your teacher will instruct you on this) and record the adjusted value in Table 1.
4 Repeat steps 1 to 3 for the 'Normal' and 'High' urine samples and record the results in Table 1. Ensure you thoroughly clean the urine hydrometer and jar between each sample.

Unknowns testing

1 Repeat the entire testing procedure for samples Unknown A and Unknown B. Note the colour and determine the pH, glucose, protein and specific gravity values of each sample. Record the results in Table 1.
2 Using Tables A and B below as reference, determine if the test results of Unknown A and Unknown B indicate any disease/s. Record your results in Table 2.

TABLE A Urine colour and possible causes

COLOUR	DIET	DRUGS	DISEASE
Light yellow to amber	Normal	None	None
Clear to light yellow	Increased fluid intake	Alcohol	Uncontrolled diabetes mellitus
Yellow-orange to orange	Carrots	Antibiotics, Pyridium	Bilirubin from obstructive jaundice
Green	Green food dyes, asparagus	Diuretics	Bacterial infection
Red to red-brown	Beets	Senna laxatives	Haemoglobin in urine (various causes)
Dark wine	Beets	Anti-inflammatory drugs	Haemolytic jaundice
Brown	Rhubarb (large quantity), fava beans, severe dehydration	Barbiturates	Haemolytic anaemia or liver disease; extremely strenuous exercise or muscle injury
Brown to black	Rhubarb (huge quantity), excessive sorbitol consumption	Antidepressants	Melanin pigment from melanoma (rare)

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TABLE B Abnormal urinalysis results and causes

TEST RESULT	DIET	DISEASE
Low pH (<6)	High protein diet; cranberry juice	Uncontrolled diabetes mellitus
High pH (>8)	Diet rich in vegetables; dairy products	Severe anaemia
Low specific gravity (<1.010)	Increased fluid intake	Severe renal damage
High specific gravity (>1.026)	Decreased fluid intake; loss of fluids	Uncontrolled diabetes mellitus; severe anaemia
Glucose present	Large meal	Uncontrolled diabetes mellitus
Protein present	High-protein diet	Severe anaemia

Studying your results

1 Copy and complete Table 1 with the results of your investigation.

TABLE 1 Simulated urine samples

URINE TEST	LOW	NORMAL	HIGH	UNKNOWN A	UNKNOWN B
Colour					
pH					
Glucose					
Protein					
Specific gravity					

2 Place an asterisk next to all abnormal results in Table 1.
3 Copy and fill out Table 2 with the results of your investigation.

TABLE 2 Unknown urine samples

SAMPLE	DISEASE INDICATED	EVIDENCE
Unknown A		
Unknown B		

Discussion

1 What information regarding patient health does examining urine under a microscope provide?
2 Describe how urinalysis results may change for an individual over a 24-hour period. Consider daily activities and how test results may be different in urine samples collected earlier or later in the day.

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3 In Table 3, describe the urinalysis results that you would expect to see from 'Patient A' and provide reasoning for each factor. Patient A has been diagnosed with strep throat and has been prescribed amoxicillin to be taken twice a day. They are under instruction to get lots of bed rest and drink plenty of fluids.

TABLE 3 Urinalysis results for Patient A

FACTOR	RESULT	REASONING
Colour		
pH		
Glucose		
Protein		
Specific gravity		

4 What side-effect warning would be appropriate for an anti-inflammatory medication used to treat the symptoms of arthritis?

5 Why do doctors order a urinalysis when attempting to diagnose diabetes? Can these urine tests be used to definitively diagnose diabetes? Discuss.

Taking it further

1 Investigate other types of urine tests, such as drug and pregnancy tests. Describe how each test works.

2 Although rare, false positive results can occur in both pregnancy and certain drug tests. This can be due to diet, medications and other factors. Describe how false positive pregnancy and drug tests occur.

3 Compare urine and blood testing. Identify the advantages and disadvantages of the two tests.

4 Investigate how urinalysis differs between animals and humans as a result of physiological differences.

ACTIVITY 6.2 Investigating breathing rate

In this activity, you will consider the stimuli that could be involved in regulating breathing rate.

You will need

Stopwatch or clock with a second hand; large paper bag

What to do

Warning: Do not act as a subject for this activity if you suffer from any respiratory or heart problems.

Work with a partner. Read through these instructions and draw up a suitable table in which to record your results.

1 Each member of the pair should count their own breathing rate (in breaths per minute) while sitting quietly at rest. Record the resting breathing rate.

2 After a normal quiet expiration, hold your breath for as long as you can. Count and record your breathing rate immediately after holding your breath.

3 Flatten a brown paper bag so that it has little air in it. Place the opening of the bag over your nose and mouth so that you are re-breathing the same air. Breathe into and out of the bag for one minute. Count and record your breathing rate immediately after breathing into the bag.

Studying your results

1. How did your breathing rate change after holding your breath and after breathing into the paper bag?
2. Suggest reasons for the changes in breathing rate.
3. What could be the stimulus that regulates a person's rate of breathing?
4. How does the evidence from the activity you have just done support your answer to Question 3?

ACTIVITY 6.3 Investigating behaviour and homeostatic mechanisms

Design an investigation to test links between one type of behaviour and one aspect of homeostasis. Some behaviours from which you could choose are:

- moderate physical exercise, such as a brisk walk or a jog
- performing relaxation exercises
- playing an exciting computer game
- watching a scary movie
- meditating
- debating a controversial topic with a friend or family member
- listening to a particular type of music – for example, relaxation music, metal or techno
- any other behaviour that you think could affect homeostasis.

Aspects of homeostasis that you could investigate are:

- blood pressure
- heart rate
- breathing
- body temperature
- any other aspect of homeostasis that you can observe or measure.

Planning your investigation

Some of the questions that you will need to answer in your planning are as follows.

- What hypothesis will you test? The hypothesis should link the behaviour you are going to test with the aspect of homeostasis that you are going to investigate. Make sure your investigation really will test your hypothesis.
- What data will you collect? How will you make your observations objective? Quantitative measurements are the best option, if possible.
- What variables will you control, and how will you go about controlling them?
- How will you make sure that your results are valid and reliable? How many repetitions will you perform?
- How will you record your results? Will it be possible to present the results as a table and/or a graph?

Conclusions

Write a conclusion discussing the relationship between your results and your hypothesis.

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CHAPTER 6 SUMMARY

- Water makes up 45–75% of the mass of the body and plays a role in transporting substances from one area of the body to another, facilitating movement across membranes and being the site of chemical reactions.
- The fluid in the body is distributed between the intracellular and extracellular fluid. Extracellular fluid is contained in plasma, between the cells, and in certain body regions such as the brain, spinal cord, eyes and joints, and surrounding the heart.
- Fluid is obtained from eating, drinking and metabolic processes, and is lost through the kidneys, skin, lungs and alimentary canal.
- Excretion is the removal of the waste products of metabolism. It is carried out by the lungs, sweat glands, alimentary canal and kidneys.
- The only water loss that can be regulated is from the kidneys.
- Water is reabsorbed from the filtrate by osmosis in the proximal convoluted tubule and loop of Henle, and by active reabsorption in the distal convoluted tubule and collecting duct.
- Antidiuretic hormone (ADH) controls the active reabsorption of water in the nephrons by increasing the permeability of the distal convoluted tubules and collecting ducts. It is produced by the hypothalamus and released from the posterior pituitary gland in response to stimulation of osmoreceptors in the hypothalamus by an increase in osmotic pressure (lower water levels).
- Aldosterone is produced by the adrenal cortex. It stimulates the reabsorption of sodium ions and the secretion of potassium ions. It also increases the reabsorption of water.
- Osmoreceptors stimulate the thirst centre in the hypothalamus when there is an increased osmotic pressure. This makes the person thirsty. When they drink, water is absorbed from the alimentary canal into the blood.
- Dehydration occurs when there is insufficient water in the body. If untreated, it can be fatal as the body cannot carry out essential functions.
- Drinking excessive amounts of water can lead to water intoxication. The body fluids become too dilute and the cells swell due to osmosis.
- Cellular respiration requires oxygen and produces carbon dioxide; therefore, the concentration of these gases needs to be regulated.
- Breathing is responsible for the input of oxygen and the excretion of carbon dioxide. It is caused by the diaphragm and intercostal muscles, which are controlled by spinal nerves.
- The respiratory centre is in the medulla oblongata. It controls and coordinates inspiration and expiration.
- Chemoreceptors detect changes in the concentration of chemicals. Peripheral chemoreceptors are found in the aortic and carotid bodies, and detect the concentration of oxygen, carbon dioxide and hydrogen ions. Central chemoreceptors are found in the medulla oblongata and detect the concentration of carbon dioxide and hydrogen ions.
- An increase in the concentration of carbon dioxide will lead to an increase in the concentration of hydrogen ions due to the reaction between carbon dioxide and water forming carbonic acid, which ionises to form hydrogen ions.
- A large decrease in oxygen is needed before the peripheral chemoreceptors are stimulated.
- A small increase in carbon dioxide will stimulate the central chemoreceptors; however, the response is relatively slow.

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- A small increase in carbon dioxide will lead to an increase in hydrogen ions that can be detected by the peripheral chemoreceptors. This initiates a rapid response.
- When chemoreceptors are stimulated, they send a message to the respiratory centre, which increases the rate and depth of breathing by the contraction and relaxation of the diaphragm and intercostal muscles.
- Breathing can also be initiated and controlled by the cerebral cortex, bypassing the respiratory centre. However, if the concentration of carbon dioxide rises, the respiratory centre will override the voluntary control, forcing the person to take a breath.
- Hyperventilation occurs when the rate of breathing is too fast. It increases the concentration of oxygen and decreases the concentration of carbon dioxide. Under normal circumstances, chemoreceptors detect these changes and reduce the urge to breathe. This can be dangerous while swimming under water, as the person may lose consciousness due to the lack of oxygen before they feel the need to breathe.
- Exercising will increase the use of oxygen and the production of carbon dioxide. Therefore, the rate of breathing will increase.

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CHAPTER 6 GLOSSARY

Aldosterone A hormone that acts on the kidney to reduce the amount of sodium in the urine and increase the amount of potassium

Antidiuretic hormone (ADH) A hormone produced by the hypothalamus and released by the posterior lobe of the pituitary gland that stimulates the kidneys to remove water from urine, thus reducing urine production; also known as vasopressin

Aortic body The group of cells within the walls of the aortic arch that are sensitive to changes in the concentrations of oxygen and carbon dioxide in the blood and its pH

Carotid body The group of cells within the walls of the carotid arteries that are sensitive to changes in the concentrations of oxygen and carbon dioxide in the blood and its pH

Chemoreceptor A receptor sensitive to particular chemicals

Cytosol The liquid part of the cytoplasm of a cell

Dehydration Excessive loss of water and accompanying salts from the body; results when the body loses more fluid than it takes in

Excretion Removal of the wastes of metabolism from the body

Extracellular fluid Fluid outside the body cells; includes tissue fluid and blood plasma

Hyperventilation Extremely rapid or deep breathing; may result in dizziness, and even fainting, due to the loss of carbon dioxide from the blood

Intercellular fluid *see* interstitial fluid

Interstitial fluid Fluid between the body cells; also known as intercellular fluid or tissue fluid

Intracellular fluid Fluid found inside cells; also known as cytosol

Intravascular fluid Fluid inside the blood vessels; also known as plasma

Kidney One of a pair of excretory organs responsible for maintaining a constant concentration of substances in the body fluids

Metabolic water Water formed as a by-product of cellular respiration

Nephron The functional unit of the kidney

Osmoreceptor A receptor sensitive to osmotic pressure of body fluids

Osmotic concentration The concentration of solutes; also known as osmolarity

Osmotic pressure The tendency of a solution to take in the pure solvent

Respiratory centre The part of the brain that regulates breathing rate; located in the medulla oblongata

Thirst centre The part of the brain that regulates the feeling of thirst; located in the hypothalamus

Tissue fluid *see* interstitial fluid

Transcellular fluid The fluid in specific body regions such as the brain, eyes and joints

Ureter The tube that leaves each kidney and drains into the urinary bladder

Urethra The tube that empties the bladder to the outside; in males, it also carries sperm

Urinary bladder A hollow, muscular organ near the base of the abdominal cavity; collects urine from the two ureters

Water intoxication A potentially life-threatening condition caused by drinking too much water when the amount of salt (and other electrolytes) in the body is low; commonly caused by long bouts of intensive exercise during which electrolytes are not replenished and large amounts of water are consumed

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CHAPTER 6 REVIEW QUESTIONS

Recall

1 State whether the nervous system, endocrine system, or both, are responsible for:
   a body fluid homeostasis
   b gas concentration homeostasis.

2 a What are nephrons?
   b Draw a diagram of a nephron and label the places where filtration, reabsorption and secretion occur.
   c Describe the role of nephrons in homeostasis.

3 Describe the role of antidiuretic hormone (ADH) in regulating water output.

4 Describe the effects of the following factors on breathing rate:
   a concentration of oxygen in the blood
   b concentration of carbon dioxide in the blood
   c hydrogen ion concentration (pH) in the blood.

5 Describe the role of the aortic and carotid bodies in regulating breathing rate.

6 Draw a pie chart to demonstrate the distribution of body fluids.

7 Define 'metabolic water'.

8 Name and describe the receptors that play a role in the control of:
   a body fluid
   b gas concentrations.

Explain

9 Explain how the respiratory centre controls the rate of breathing.

10 Aldosterone regulates the amount of sodium in the blood. Explain why aldosterone influences the amount of water excreted from the body.

11 We cannot voluntarily control our heart rate or blood sugar level, yet we can voluntarily control our breathing.
   a Explain why it is important for us to be able to voluntarily decide when to take a breath and how deep the breath should be.
   b We cannot voluntarily stop breathing indefinitely. Explain why.

12 Explain why excretion is closely related to maintaining fluid balance.

Apply

13 A person lost in the desert would suffer extreme dehydration. Although the thirst receptors would try to initiate drinking behaviour, the lack of available water would not allow this requirement to be met. Describe the mechanisms the body would employ to conserve water while getting rid of metabolic wastes.

14 A student made the following observations. On a very hot day, the volume of urine produced was small and it was dark in colour. On a cold day, urination occurred more frequently, and the urine was pale in colour. Explain these observations.

15 An athlete had blood samples taken before and after a vigorous training session on a hot, dry day. The sample taken after training had a much higher concentration of ADH than the sample taken before training. Explain why there would be a difference in concentrations.

16 Moderate dehydration occurs when the body loses 7–10% of the body weight in fluid. What is the minimum mass an 80 kg male would need to lose in fluid to be considered moderately dehydrated?

17 Why is it dangerous to hyperventilate before swimming under water?

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18 People sometimes hyperventilate in stressful situations. The hyperventilation may cause dizziness and tingling of the fingers and toes. In such cases, the person may be advised to breathe into a paper bag and re-breathe the same air that was breathed out. How would such a procedure help to overcome the problems of hyperventilation?

19 Draw a stimulus–response–feedback diagram, labelling the receptor, modulator, effector and feedback, to show what happens to breathing rate when:

- **a** the concentration of carbon dioxide in the blood increases
- **b** the hydrogen ion concentration of the blood increases.

Extend

20 Use a flow chart to summarise all of the changes that would occur in the body while playing basketball during a hot day.

21 The table shows the water loss from a person's skin and kidneys under different conditions. Use the data to explain the relationship between regulation of body temperature and regulation of fluid content of the body.

22 Elderly people are much more likely to suffer from water regulation problems than the young. The gradual decline in the effectiveness of the thirst reflex has already been mentioned, but there is also a decline in the effectiveness of the kidneys. Find out the changes in kidney function that occur with age, and describe some of the conditions that can occur in elderly people due to poor kidney function.

ORGAN	WATER LOST (mL/HOUR)		
	AT ROOM TEMPERATURE	IN HOT WEATHER	WITH LENGTHY VIGOROUS EXERCISE
Skin	19	73	225
Kidneys	58	50	20

7 THE BODY CAN PROTECT ITSELF FROM INFECTION

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

- identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes
- design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics, including animal ethics
- conduct investigations, including the collection of data related to homeostasis and the use of models of disease transmission, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports

SCIENCE AS A HUMAN ENDEAVOUR

- the decision to participate in immunisation programs can be influenced by the social, economic and cultural context in which it is considered

SCIENCE UNDERSTANDING

Response to infection

- infectious diseases caused by invasion of pathogens in the form of viruses and bacteria can be transmitted from one host to another
- transmission of pathogens occurs by various mechanisms, including through:
  - direct and indirect contact
  - transfer of body fluids
  - disease-specific vectors
  - contaminated food and water
- the body's external defence mechanisms against pathogens include features of the:
  - skin
  - digestive tract
  - urogenital tract
  - respiratory system
  - the ear
  - the eye
- pathogens that enter the body are targeted by non-specific immune responses of inflammation and fever
- immunity is gained through the exposure to specific antigens by the production of antibodies by B lymphocytes and the provision of cell-mediated immunity by T lymphocytes; in both cases memory cells are produced
- passive immunity can be acquired as antibodies gained through the placenta, or antibody serum injections; active immunity can be acquired through natural exposure to the pathogen, or the use of vaccines
- antiviral and antibiotic drugs are used for treating infections and differ in their specificity to pathogens

Source: School Curriculum and Standards Authority, Government of Western Australia

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The human body has a number of mechanisms to protect it from invading organisms. If the body's defences are overcome, the invaders may cause disease. Such disease-causing organisms are called **pathogens**. Some diseases are spread from one person to another. These diseases are **communicable**, or **infectious**, **diseases** and are also called transmissible diseases.

We are often exposed to pathogens without realising it. Luckily, our bodies have a number of defences that protect us from them. Many pathogens are prevented from entering the body or, if they do enter, are dealt with before they can cause symptoms of disease. Even if we do become ill, the body's defence system often enables recovery without any medical intervention.

7.1 PATHOGENS

The most common pathogens that affect the human body are bacteria and viruses, although fungi and animal parasites can also be involved. In this chapter, we will be focusing on bacteria and viruses.

Bacteria

Bacteria are **prokaryote**, unicellular organisms with a simple internal structure. They lack a nucleus; their DNA either floats freely in the cytoplasm or is in the form of circular **plasmids**.

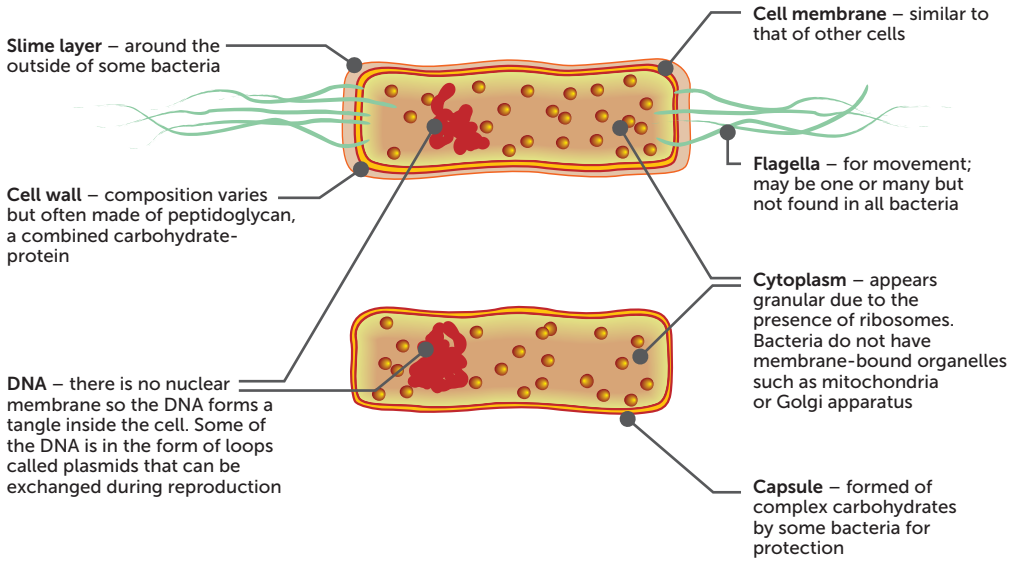

FIGURE 7.1
Structure of a typical bacterial cell

The great majority of bacteria are harmless to humans; they are non-pathogenic. Indeed, many bacteria are essential to life on Earth, through their role in the decomposition of organic material and the cycling of the elements. Some bacteria are used in industrial processes. For example, *Lactobacilli* are used to make yoghurt and sauerkraut; and the flavour of cheeses depends on the types of bacteria used in their production.

Huge numbers of bacteria live on our skin, in our alimentary canal and in other parts of the body. In the armpit of an adult male, there are more than two million bacteria per square centimetre of skin surface; and in the intestines, bacteria are so numerous that they form a major part of the digestion process. These bacteria have no ill effect on our health, yet there are others that may cause illness or death when present in relatively small numbers. Bacteria affect the body differently, depending on the species. Effects may include producing toxins or inducing an allergic response.

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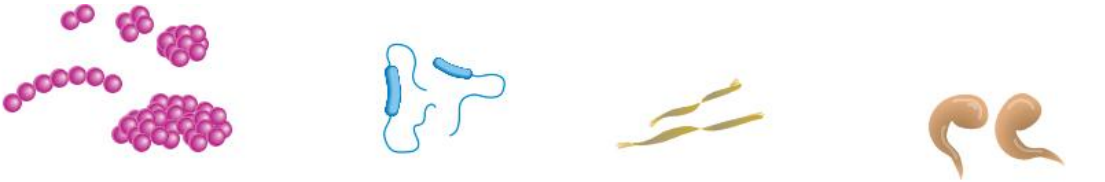
FIGURE 7.2
Types of bacteria, classified according to cell shape



Cocci (singular 'coccus') are spherical cells that may occur singly, in pairs (diplococci), in clusters (staphylococci) or in chains (streptococci)

Bacilli (singular 'bacillus') with **flagella** have rod-shaped cells; many have flagella for movement

Spirilla (singular'spirillum') have twisted cells

Vibrio are like curved rods and are often shaped like a comma

Bacteria are very small, with the average diameter ranging from 0.5 to 2.0 μm (micrometres; 1 μm = 1 x 10^{-6} m) and length ranging from 1 to 10 μm. This means that bacteria can be seen only with a microscope. Under the light microscope, about all that can be seen of bacteria is the shape of their cells, which is used to classify them.

To identify a bacterium, it is first grown on an agar plate or growth medium in specific conditions. Then it can be stained and viewed under a microscope.

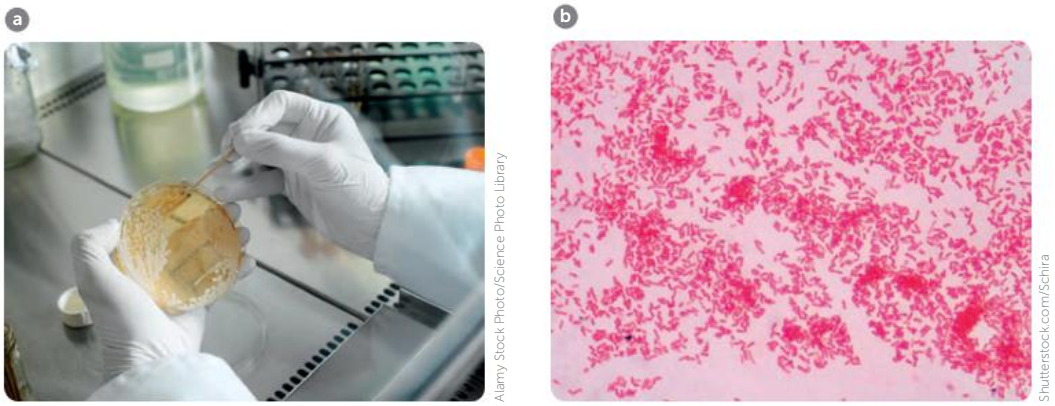


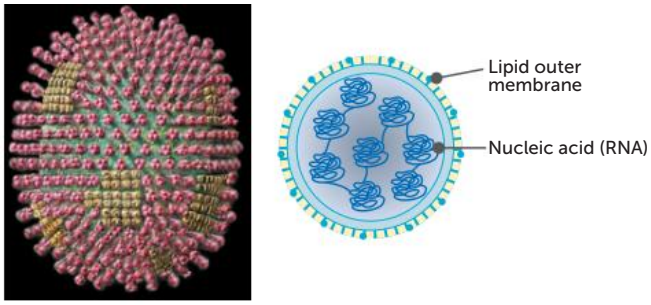
FIGURE 7.3 a Bacteria that have invaded the body can be cultured. **b** Bacteria can be viewed under a microscope as part of their identification. Some bacteria are pink when using a gram stain technique, while others are purple.

Viruses

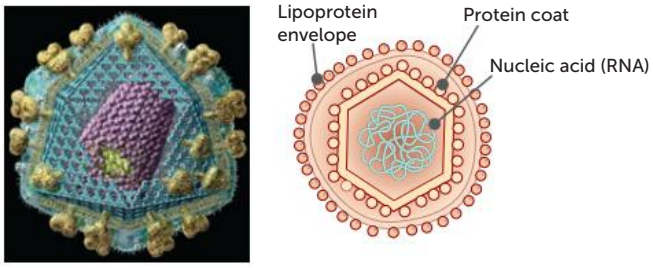
Activity 7.1
Investigating the effectiveness of hand washing

The discovery, by scientists such as Pasteur and Koch in the late 19th century, that some diseases were caused by bacteria was a great step forward for medical science. There were, however, certain diseases for which no bacterial cause could be found. For example, Pasteur tried in vain to find a bacterium that caused the disease rabies. We now know that the causes of these diseases are **viruses**.

Viruses are from 20 to 750nm (nanometres; 1 nm = 1 x 10^{-9} m) in size, which is too small to be seen with an ordinary light microscope. It wasn't until 1938 that scientists used an electron microscope to first see viruses. Subsequent studies showed that viruses had distinctive structures and differing sizes. All were found to contain genetic material in the form of a molecule of either DNA (deoxyribonucleic acid) or RNA (ribonucleic acid), but they never contained both. The molecule of DNA or RNA is surrounded by a coat of protein. Some viruses also have an external lipid envelope.



Influenza virus has a lipid outer membrane. The RNA is in eight segments.

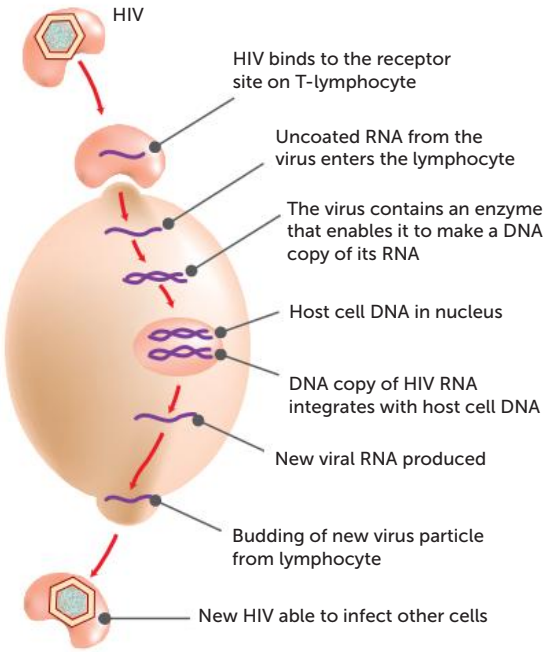


Human immunodeficiency virus (HIV) has a lipoprotein envelope with an internal protein coat.

Viruses all contain either DNA or RNA but not both. Around the nucleic acid is a protein coat, and some viruses have an additional envelope of lipid and protein molecules.

FIGURE 7.4
Structure of the influenza virus and human immunodeficiency virus

Viruses are not living things, as they cannot reproduce by themselves. Instead, they infect a living cell and its DNA or RNA induces the cell to manufacture more virus particles. The new virus particles are then able to leave the host cell to infect others. During this process the cells become damaged or changed, or die. Viruses differ in the type of cell they invade; therefore, the symptoms shown relate to the tissue that is affected. Some viruses multiply in bacterial cells, causing the death of the bacterium. Such viruses are known as **bacteriophages**.



Viruses cannot reproduce themselves. They attach to the outside of a host cell and the nucleic acid enters the cell. New viral genes are produced by the host cell, and so hundreds of new virus particles are formed.

FIGURE 7.5
The process of viral replication illustrated by HIV

Viruses
Khan Academy has more information about viruses.

ViralZone
Click on the name of the virus to view its structure.

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TABLE 7.1 Some of the better-known diseases caused by pathogens

BACTERIA	VIRUSES	FUNGI	ANIMAL PARASITES
Anthrax	HIV/AIDS	Ringworm	Protozoans
Botulism	Bird flu	Thrush	Amoebic dysentery
Bubonic plague	Chickenpox	Tinea	Amoebic meningitis
Chlamydia	Cold sores (herpes)		Malaria
Cholera	Colds		Sleeping sickness
Dental caries (tooth decay)	COVID-19		Toxoplasmosis
Diphtheria	Ebola		Platyhelminthes (flatworms)
Gastroenteritis	Encephalitis (viral)		Blood flukes
Gonorrhoea	Genital herpes		Hydatids
Impetigo (school sores)	Glandular fever		Liver flukes
Legionnaire's disease	Hepatitis A, B, C, D, E and G		Tapeworms
Leprosy	Influenza		Nematodes (round worms)
Meningitis (bacterial)	Measles		Hookworms
Peptic ulcers	Meningitis (viral)		Roundworms
Pneumonia	MERS (Middle East respiratory syndrome)		Threadworms
Scarlet fever	Mumps		Arthropods
Syphilis	Poliomyelitis		Lice
Tetanus	Rabies		Scabies (mites)
Trachoma	Ross River virus		Ticks
Tuberculosis	Rubella		
Typhoid	SARS (severe acute respiratory syndrome)		
Whooping cough	Shingles		
	Smallpox		
	Warts		
	Yellow fever		

Transmission of pathogens

Communicable disease may be spread by the transmission of the pathogenic organism from one person to another. Some communicable diseases are said to be **contagious**; this means they are passed directly from one person to another. Other communicable diseases may be spread from person to person by **vectors**; intermediate hosts of the pathogen, such as mosquitoes or fleas.

Transfer can occur in a number of ways.

- *Transmission by contact* involves the spread of the pathogen by actual physical contact. The contact may be *direct*, actually touching an infected person; or *indirect*, touching an object that has been touched by an infected individual. Skin infections and sexually transmissible infections are spread by contact.
- *Ingestion* of food or drink contaminated with pathogens may result in disease. Dysentery, typhoid fever and *Salmonella* food poisoning are transmitted in this way.
- *Transfer of body fluids* from one person to another can result in the transmission of a number of infections. When blood or other body fluids from an infected person comes into contact with the mucous membranes, such as in the nose, mouth, throat and genitals, or the bloodstream of an uninfected person, such as through a needle stick or a break in the skin, then pathogens may enter the body of that person. The human immunodeficiency virus, and hepatitis B and C, are spread in this way.
- *Infection by droplets* may occur when tiny droplets of moisture containing pathogenic organisms are emitted when breathing, talking, sneezing or coughing. The droplets may be breathed in by others, or may settle on food or utensils to be later ingested with food. Many viral infections, such as those causing Ebola, COVID-19, mumps, colds and influenza, can be spread by droplets.

Ebola in the air
This website has more information about the spread of the Ebola virus, and transmission by droplets and airborne particles.

- *Airborne transmission* of some diseases may occur. When the moisture in exhaled droplets evaporates, many bacteria are killed, but viruses and some bacteria remain viable and can cause infection when inhaled. As these particles are lighter, they remain viable for a greater distance than those transmitted by droplets. Measles and chickenpox are spread by this method.

Transmission of measles

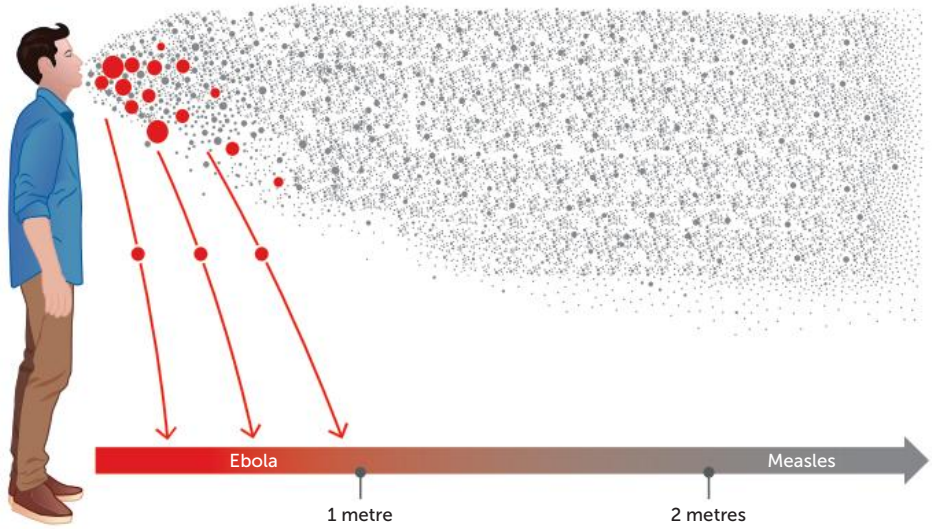


FIGURE 7.6
Transmission of pathogens by droplets (red) and airborne (grey) routes

- *Transmission by vectors* is the transfer of pathogens by other animals, such as insects, ticks or mites. Some vectors transfer the pathogen directly; others, such as house flies, may spread the pathogen to food or water, which is then ingested. Many vector-borne diseases are spread by a specific vector. For example, malaria and dengue fever are spread by mosquitoes, trypanosomiasis (African sleeping sickness) is spread by the tsetse fly, Lyme disease is spread by ticks, and bubonic plague is spread by fleas from rats and mice.

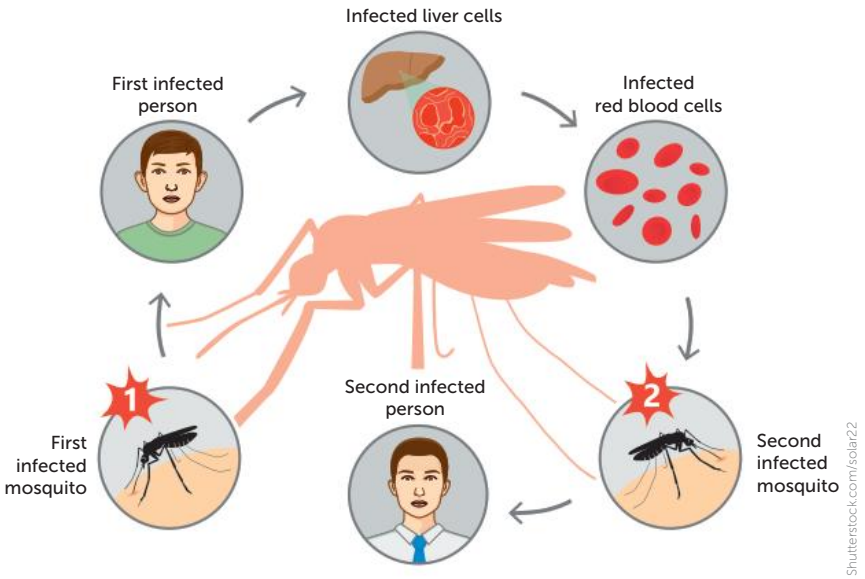


FIGURE 7.7
Transmission of malaria via a mosquito vector

Key concept

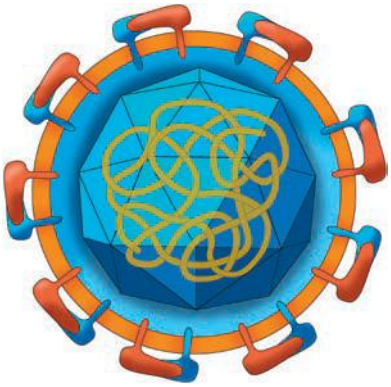
Communicable diseases are caused by pathogens such as bacteria and viruses and are spread from person to person, either directly or indirectly.

Activity 7.2
Investigating infectious disease transmission

Questions 7.1

RECALL KNOWLEDGE

1 Define 'pathogen'.
2 List the ways that a pathogen can pass from an infected person to someone else.
3 The diagram below is of the zika virus. Label the nucleic acid, protein capsule and lipid envelope.



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4 Describe the structure of a typical bacterium.
5 List the different shapes of bacteria.
6 List three vectors of pathogens and the diseases that they transmit.

APPLY KNOWLEDGE

7 During the COVID-19 pandemic, people were advised to wash their hands with soap to break down the coronavirus's protein coat. Explain how this would be effective in preventing the transmission of the virus.
8 There is some debate as to whether viruses are living things. Discuss your views on this idea.
9 It is easier to stop the transmission of pathogens that are transferred by body fluids than those that are transferred by moisture. Explain the reason for this observation.

7.2 NON-SPECIFIC DEFENCES AGAINST DISEASE

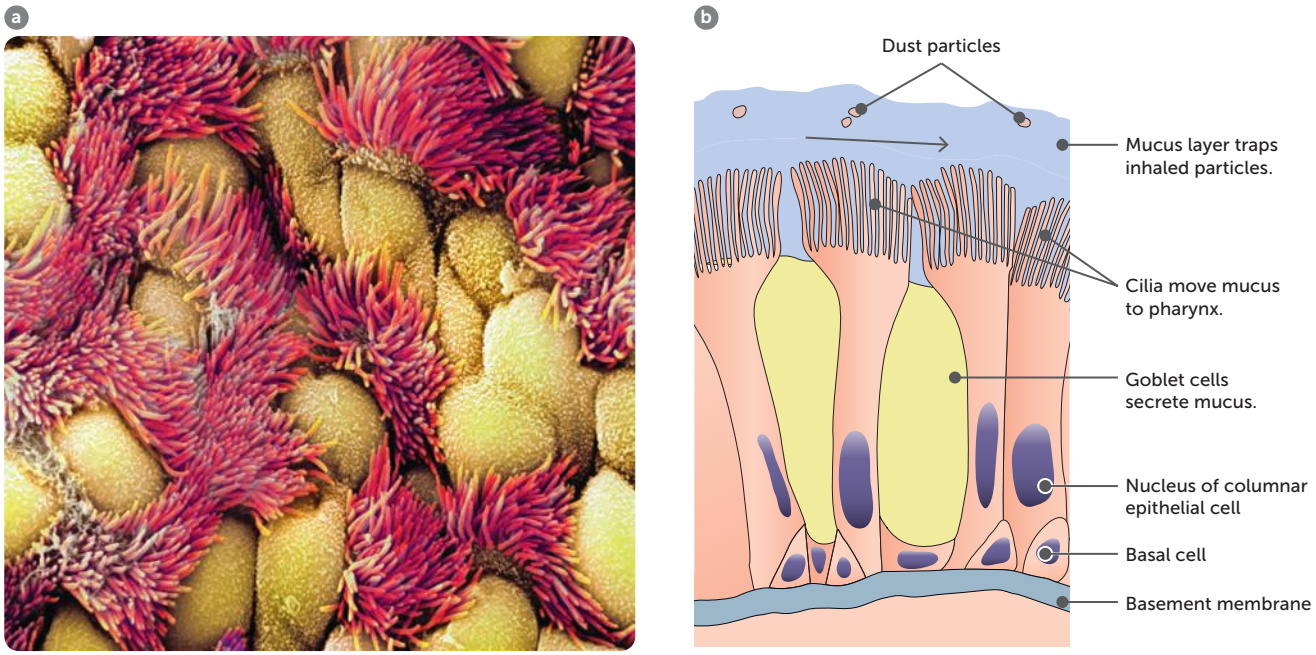
Our bodies have several defences that protect us against invasion by pathogenic micro-organisms. We are often exposed to pathogens without realising it. Many pathogens are prevented from entering the body or, if they do enter, they are dealt with before they can cause symptoms of disease. Even if we do become ill, our defence system often enables us to recover without any medical intervention.

The body's defences can be classified as specific or non-specific based on what pathogens it works against. **Non-specific defences** work against all pathogens. They are the body's first line of defence. **Specific defences** are directed at a particular pathogen.

External defences

The body has many external defences to try to stop pathogens, and other foreign particles, from entering. These are all non-specific. Some of the external defences are as follows.

- *Skin*: The skin is an effective barrier covering the outside of the body. It is very good at stopping the entry of micro-organisms, provided it is not broken by cuts and abrasions. At openings in the skin, such as the mouth, eyes and anus, special protection is provided by other defences. Huge numbers of bacteria live on the skin all the time. These normal bacteria occupy the area, and so potential pathogens find it difficult to become established. In addition, the skin has other protective mechanisms. An oily secretion called **sebum** is produced by oil glands in the skin. It contains substances that kill some pathogenic bacteria. **Sweat** secreted on to the skin contains salts and fatty acids that prevent the growth of many micro-organisms.
- *Mucus*: **Mucous membranes** line body cavities that open to the exterior. They secrete **mucus**, which traps particles and, therefore, inhibits the entry of micro-organisms to the organs of the body. The digestive, urinary and reproductive tracts are all protected in this way.
- *Hairs*: Hairs are found in the **nasal cavity** in the nose, and in the ears. In the nose, the hairs and a layer of mucus trap up to 90% of particles inhaled when breathing.
- *Cilia*: **Cilia** are tiny hair-like projections from cells that are capable of a beating motion. The mucous membranes lining the nasal cavity, the trachea and other air passages have cilia. The beating of the cilia moves mucus, containing trapped particles and micro-organisms, towards the throat, where it may be coughed up or swallowed.
- *Acids*: Stomach juices are strongly acidic. The acid kills many of the bacteria taken in with food or those contained in mucus swallowed from the nose and windpipe. The vagina also has acid secretions that reduce the growth of micro-organisms. Urine and the sweat on the skin are also slightly acidic.



Science Photo Library/NIBSC

FIGURE 7.8 a Scanning electron micrograph showing the cilia of cells lining the respiratory system; the structures between the cilia are mucus-secreting cells. **b** Mucus traps foreign particles and cilia move it out of the body.

- *Lysozyme*: **Lysozyme** is an enzyme that kills bacteria. The eyes are protected by the flushing action of tears, which contain this enzyme. Lysozyme is also found in saliva, sweat, secretions of the nose and tissue fluid.
- *Cerumen*: **Cerumen**, or ear wax, protects the outer ear against infection by some bacteria. It is slightly acidic and contains lysozyme.
- *Movement of fluid*: The flushing action of body fluids helps to keep some areas relatively free of pathogens. Urine flowing through the urethra has a cleansing action. This prevents bacterial growth and helps to stop bacteria reaching the bladder and kidneys. Women have a shorter urethra than men and so they tend to suffer more bladder infections. Tears, sweat and saliva are also involved in flushing and cleansing.

The body's external defences are summarised in Figure 7.9.

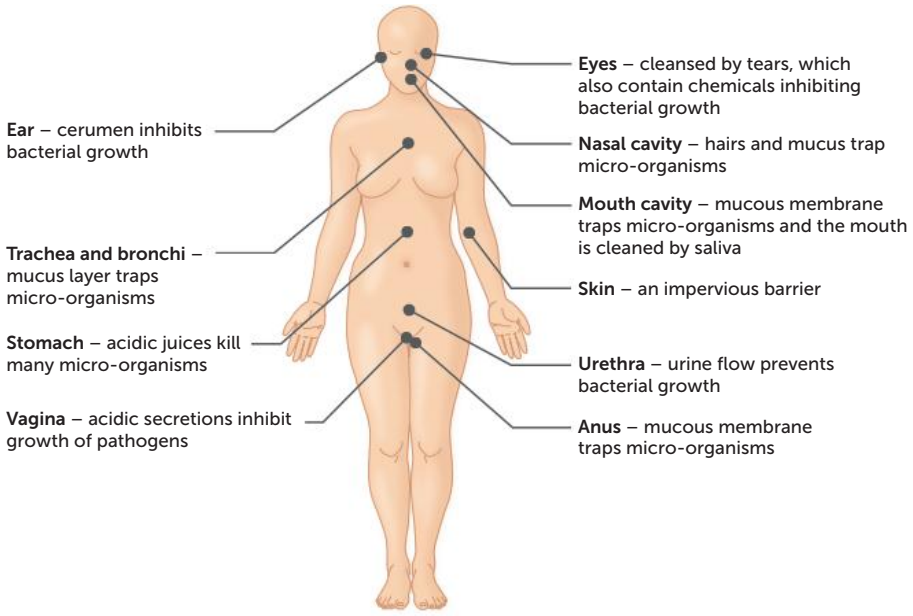


FIGURE 7.9
The body's external defences against entry of pathogenic micro-organisms

Defences against infection
This website provides an explanation of the external defences against disease.

7.1 Pathogens and non-specific immunity

Key concept

The external surfaces of the body have defence mechanisms to prevent the entry of pathogens.

Protective reflexes

A reflex is an automatic, involuntary response to a stimulus. Protective reflexes help to protect the body from injury, such as the blink reflex, or from infection, such as vomiting. Four reflexes help to protect against infection.

1. *Sneezing*: The stimulus for sneezing is irritation of the walls of the nasal cavity. The irritation may be caused by noxious fumes or dust particles, which are likely to be carrying micro-organisms. Forceful expulsion of air from the lungs carries mucus, foreign particles and irritating gases out through the nose and mouth.
2. *Coughing*: For coughing, the stimulus is irritation in the lower respiratory tract – the bronchi and bronchioles. In a manner similar to sneezing, air is forced from the lungs to try to remove the irritant. The air drives mucus and foreign matter up the trachea towards the throat and mouth.
3. *Vomiting*: Psychological stimuli, excessive stretching of the stomach and bacterial toxins can all induce vomiting. Contraction of the muscles of the abdomen and the diaphragm, not the contraction of the stomach, expels the stomach contents.

4 *Diarrhoea*: Irritation of the small and large intestines by bacteria, viruses or protozoans can cause diarrhoea. The irritation causes increased contractions of the muscles of the wall of the intestines so that the irritant is removed as quickly as possible. Material does not stay in the large intestine long enough for water to be absorbed, so the faeces are very watery.

Key concept

Protective reflexes such as sneezing, coughing, vomiting and diarrhoea remove foreign particles in an automatic, involuntary response.

Internal non-specific defences

If pathogens get past our external defences, there are internal non-specific defences that work to eliminate them.

Phagocytosis

Organisms that penetrate our external defences are attacked by phagocytes. **Phagocytes** are specialised white blood cells, or **leucocytes**, that engulf and digest micro-organisms and cell debris. This eliminates many pathogens before an infection has a chance to take hold.

There are a number of different types of cells that are phagocytic.

1 *Monocytes and macrophages*: When a tissue becomes infected or inflamed, **monocytes** leave the bloodstream and enter the tissue. Here they differentiate into **macrophages**, which are large phagocytic cells. Some macrophages move through the tissues looking for and destroying pathogens. Others are fixed in one place and only deal with the pathogens that come to them. Macrophages are particularly important in removing microbes and dying cells through phagocytosis.

2 *Neutrophils*: **Neutrophils** are described as a granulated leucocyte, due to the granules visible in their cytoplasm. They are also characterised by their lobulated nucleus.

Neutrophils are the most abundant leucocyte, accounting for 55–70% of all leucocytes. During an infection, neutrophils are the first cells to move into the tissue to destroy the pathogen by phagocytosis. They are particularly important in killing pathogens inside cells.

Neutrophils have a short life span and die after a few days. The dead cells make up a large portion of the pus that forms after an infection.

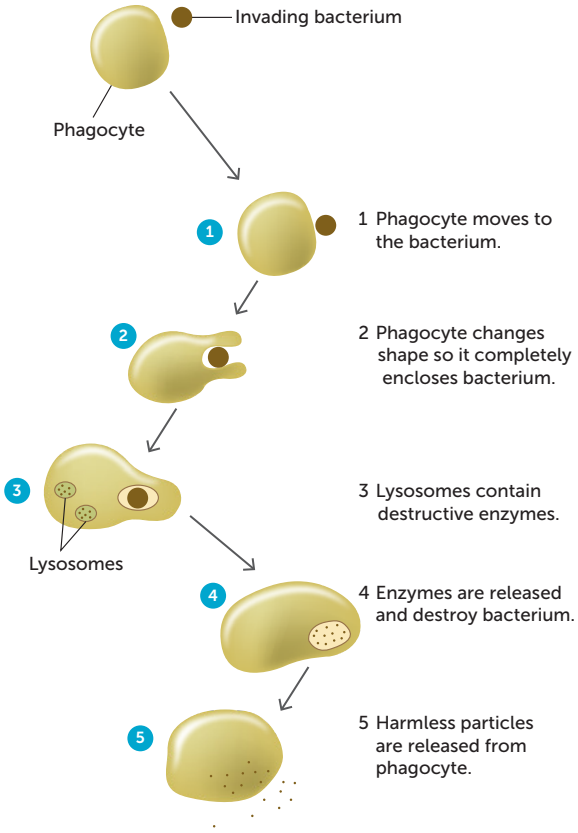


FIGURE 7.10 The process of phagocytosis

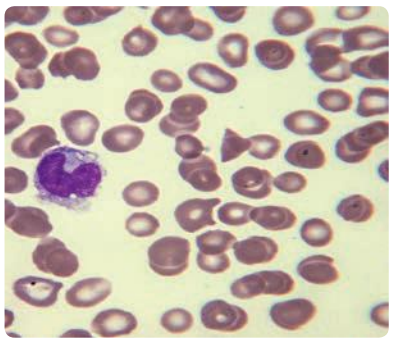
Phagocytosis
This website provides an animation of phagocytosis.

Licking wounds helps healing
This article explains why licking a wound helps it to heal faster.

Phagocytes
This website has further information about phagocytosis.

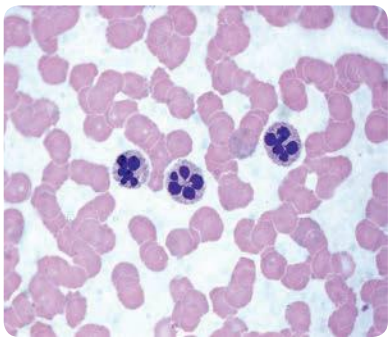
Types of leucocytes
These websites contain information about the different types of leucocytes and their functions.

3 *Dendritic cells*: **Dendritic cells** are characterised by projections from the cytoplasm. They are slightly different from macrophages and neutrophils in that their function goes beyond just phagocytosis. These cells have the ability to detect, engulf and process foreign particles. They then use information about the ingested particles to assist with specific immunity. You will learn more about dendritic cells later in this chapter.



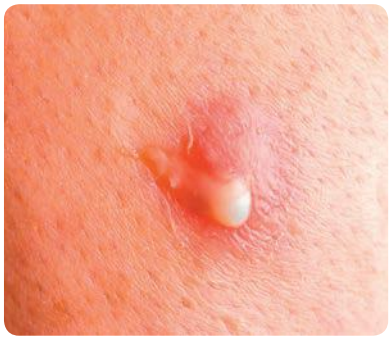
Science Photo Library/Nature's Faces/Science Source

FIGURE 7.11 Monocytes have a kidney bean-shaped nucleus



iStock.com/Jose Luis Calvo Martin & Jose Enrique Garcia-Maurino MUZQUIZ

FIGURE 7.12 Neutrophils have a granular cytoplasm and a lobulated nucleus



Shutterstock.com/Engineer studio

FIGURE 7.13 Pus forms following an infection

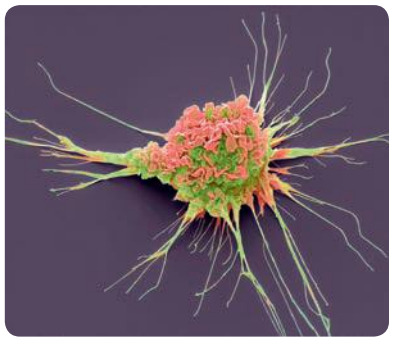


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FIGURE 7.14 Scanning electron micrograph (SEM) showing a dendritic cell with its extensions from the cytoplasm

Key concept

Macrophages, neutrophils and dendritic cells are phagocytes that engulf and destroy debris and pathogens.

Inflammatory response

Inflammation
This website provides an animation of the process of inflammation.

Words ending in *-itis* indicate **inflammation** of specific organs or tissues. For example, tonsillitis is inflammation of the tonsils; meningitis is inflammation of the meninges, the membranes around the brain; and laryngitis is inflammation of the larynx.

Inflammation is a response to any damage to the tissues. The purpose of inflammation is to:

- reduce the spread of any pathogens, to destroy them and to prevent the entry of additional pathogens
- remove damaged tissue and cell debris
- begin repair of the damaged tissue.

There are four signs of inflammation. If you think of an infected cut, a pimple or a mosquito bite, you will be able to identify each of the four signs of redness, swelling, heat and pain.

Damage to tissues stimulates a series of steps in the inflammatory response. Some of these steps are assisted by proteins in the **complement system** that are produced by liver cells and macrophages. The complement system is a series of more than 20 proteins, many of which are normally inactive. When initiated, one protein activates the next, which activates the next, and so on.

The steps of the inflammatory response include:

1. Mechanical damage or local chemical changes cause specialised leucocytes called **mast cells** to be activated by complement proteins. This results in the release of histamine, heparin and other chemicals into the tissue fluid.
2. **Histamine** increases blood flow through the area due to **vasodilation**, making the walls of the blood capillaries more permeable. More fluid moves through the capillary walls into the tissue. The increased blood flow causes heat and redness, and the escape of fluid from the blood causes swelling.
3. **Heparin** prevents clotting, so the release of heparin from the mast cells prevents clotting in the immediate area of the injury. A clot of the fluid forms around the damaged area, which slows the spread of the pathogen into healthy tissues.
4. Complement system proteins and some chemicals released by the mast cells attract phagocytes, particularly neutrophils, which actively consume micro-organisms and debris by phagocytosis.
5. The abnormal conditions in the tissue stimulate pain receptors, and so the person feels pain in the inflamed area.
6. The phagocytes, filled with bacteria, debris and dead cells, begin to die. The dead phagocytes and tissue fluid form a yellow liquid called pus.
7. New cells are produced by mitosis, and repair of the damaged tissue takes place.

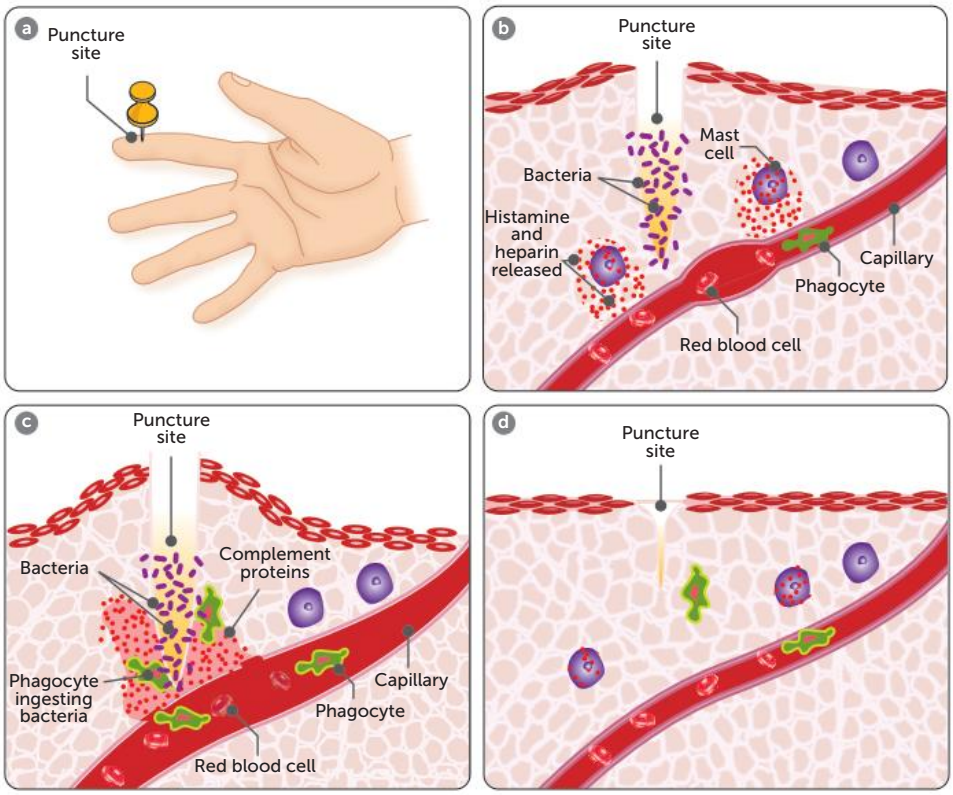


FIGURE 7.15 The inflammatory response: **a** When the skin is broken, a non-specific inflammatory response is triggered. **b** Mast cells release histamine and heparin. Histamine diffuses into capillaries, causing them to dilate and become leaky. The area becomes red and swollen. Heparin prevents clotting in the immediate area. **c** Complement proteins are activated and, along with chemicals from the mast cells, attract phagocytes to the area, which engulf and digest dead cells and bacteria. **d** The tissue heals when histamine and protein signalling finish and phagocytes are no longer attracted to the area

Key concept

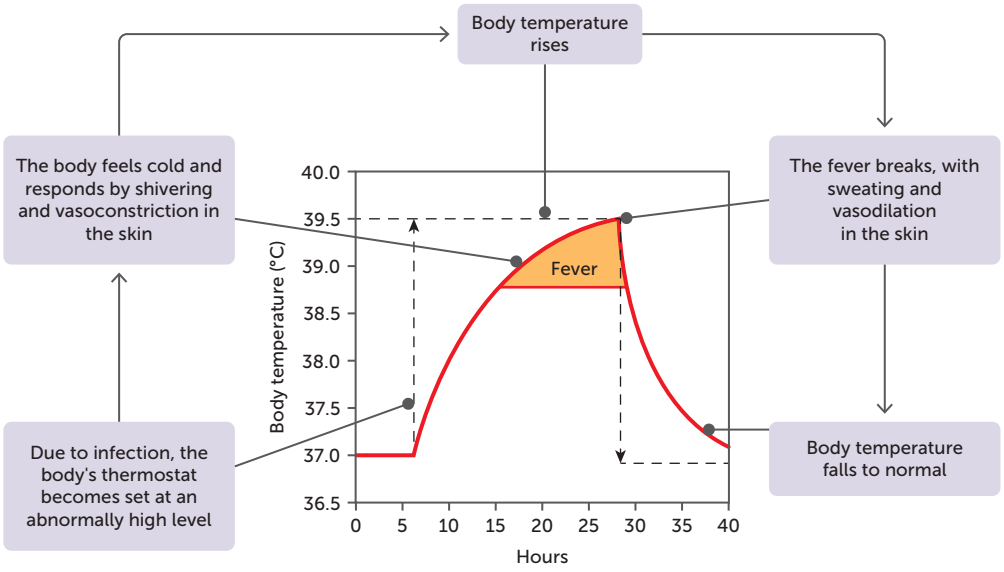
Inflammation occurs when tissue is damaged or infected. Increased blood flow, blood vessel permeability and phagocytosis lead to heat, redness, swelling and pain.

Fever

An infection such as the common cold or influenza is frequently accompanied by an elevation of body temperature, often called a **fever**. The change in body temperature is due to a resetting of the body's thermostat, controlled by the hypothalamus, to a level higher than normal. This reaction is thought to be due to chemicals called **pyrogens** that are released by white blood cells during the inflammatory response and act on the hypothalamus. One pyrogen is **interleukin-1**. It is predominantly produced by activated macrophages, but is also produced by other cells such as dendritic and epithelial cells. When a person has a fever, the body temperature is still regulated in response to heat or cold, but the set point is at a higher level.

The onset of fever is frequently gradual, but it is most striking when it occurs rapidly. In these cases, it is as though the body's thermostat were suddenly raised. The person's thermoreceptors detect the body temperature, and the hypothalamus recognises that it is lower than the new, higher set point. As a consequence, vasoconstriction in the skin and shivering occur. Both these activities conserve heat and increase heat production, driving the body temperature up rapidly. When the fever breaks, the point called the crisis, it is as though the body's thermostat has been reset to normal. In this situation the person feels hot and appears flushed, as skin vasodilation and profuse sweating take place.

FIGURE 7.16
Body temperature during fever



Fever is beneficial, up to a point. High body temperature is believed to inhibit the growth of some bacteria and viruses. In addition, heat speeds the rate of chemical reactions, which may in turn help body cells repair themselves more quickly during a disease. Fever may also inhibit viral replication by allowing chemicals called **interferons** to operate more quickly. However, if the body temperature goes too high it can cause convulsions and brain damage. Generally speaking, death will result if the body temperature reaches 44.4–45.5°C.

Activity 7.3
Plotting a fever

Key concept

Fever occurs when the body's set point for temperature is increased due to pyrogens acting on the hypothalamus. Fever can help fight infections but is dangerous if it goes too high.

Lymphatic system

The **lymphatic system** consists of:

- a network of lymph capillaries joined to larger lymph vessels
- lymph nodes, which are located along the length of some lymph vessels.

The main function of the lymphatic system is to collect some of the fluid that escapes from the blood capillaries and return it to the circulatory system. In addition to this main function, the lymphatic system is an important part of the body's internal defence against pathogenic organisms. The lymphatic system was discussed in Chapter 5 of *Human Perspectives ATAR Units 1 & 2*.

Lymph nodes occur at intervals along the lymphatic vessels. Each node contains masses of lymphoid tissue, the cells of which are criss-crossed by a network of fibres. Lymph entering the lymph nodes contains cell debris, foreign particles and micro-organisms that have penetrated the body's external defences. Some of these micro-organisms may be pathogenic and, if not destroyed, could cause disease. Larger particles, such as bacteria, are trapped in the meshwork of fibres as the lymph flows through the spaces in the nodes. Macrophages ingest and destroy these particles by phagocytosis.

When infections occur, the formation of lymphocytes increases, and the lymph nodes become swollen and sore. For example, an infected finger may result in swelling and tenderness in the armpit, where there are a large number of lymph nodes. Most lymphocytes are important in the specific immune response to a particular pathogen.

Questions 7.2

RECALL KNOWLEDGE

1. Define 'non-specific defences'.
2. Explain why the skin is such an effective external defence to infection.
3. Describe the role of cilia in the external defence mechanisms.
4. List the parts of the body that use acids to protect against disease.
5. List five protective reflexes that protect against disease or injury.
6. Name three cells that are phagocytes in tissue.
7. Draw a series of diagrams to show phagocytosis.
8. List the signs of inflammation.
9. Which cells release heparin and histamine following tissue damage?
10. Define 'fever' and 'pyrogen', and explain their relationship.
11. Name one pyrogen.
12. What is the benefit of fever during an infection?
13. Describe how lymph nodes provide non-specific defence against disease.

APPLY KNOWLEDGE

14. Explain why it is common to cough after being in a dusty environment.
15. Explain why the incidence of urinary tract infections is higher in females than in males.
16. Suggest what nephritis is.
17. Compare and contrast macrophages and neutrophils.
18. Explain how histamine causes swelling during inflammation.
19. When people are sick, they often feel cold even though their body temperature may be above normal. Explain why this happens.
20. During the COVID-19 pandemic, it was compulsory in many places to wear a mask when in public. Explain how a mask could reduce the transmission of the virus.
21. Traditionally, it was common practice to cover your mouth with your hand when coughing. Recently, it is recommended that you cough into your elbow or shoulder. Explain why this method could be more beneficial in preventing the transmission of disease.

7.3 SPECIFIC DEFENCES AGAINST DISEASE

Specific defences are those directed towards a particular pathogen. For example, if you get infected (or vaccinated) with chickenpox virus, the body will make antibodies to combat that virus. Those antibodies are only effective against chickenpox virus and will not work against any other virus or bacterium.

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Specific defences are part of our immune system. The **immune system** is composed of cells and proteins that protect against foreign organisms, a range of alien chemicals, as well as cancerous and other abnormal cells. Some of these cells are non-specific, such as phagocytes, which are able to engulf and digest micro-organisms and cell debris. However, others such as B-cells and T-cells only provide protection against a specific micro-organism or disease-causing substance. When these cells react, it is called the **immune response**.

Immune response

The immune response is a homeostatic mechanism. When micro-organisms or foreign substances enter the body, the immune response helps to deal with the invasion and restore the internal environment to its normal condition.

The key cells involved in the immune response are **B-cells** and **T-cells**, which are white blood cells called **lymphocytes**. B-cells and T-cells are both produced in the bone marrow and end up in the **lymphoid tissue**. However, they mature by different routes. About half the cells produced by the bone marrow go to the thymus, where they mature into T-cells before being incorporated into the lymphoid tissues. The other half of the cells mature in the bone marrow to become B-cells and then become part of the lymphoid tissues. Most of the lymphoid tissue is in the lymph nodes; however, it also occurs in other parts of the body, such as the spleen, thymus gland and tonsils.

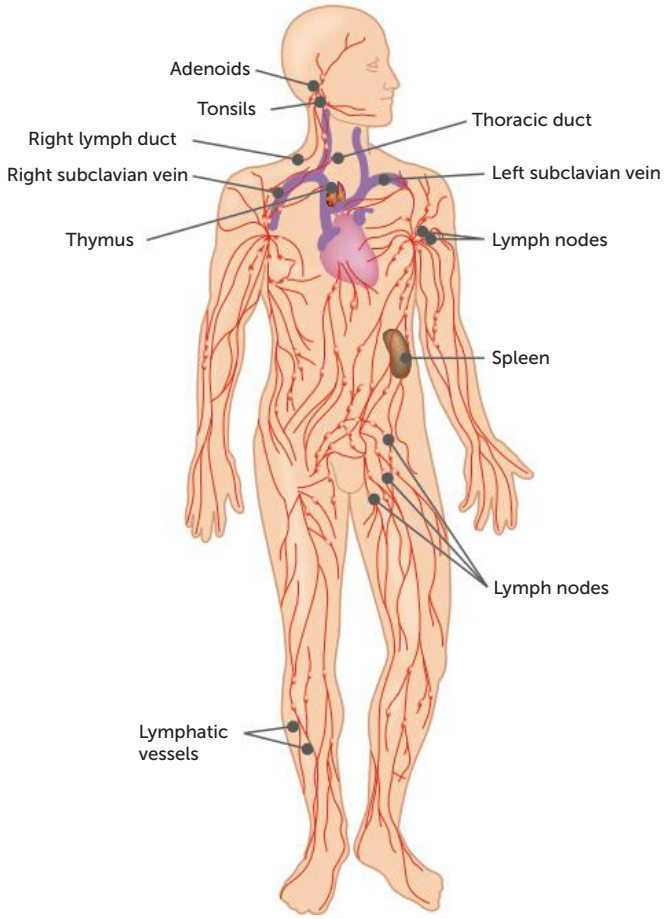

FIGURE 7.17
Lymphoid tissue occurs throughout the body

There are two parts to the immune response.

- The **humoral response** or **antibody-mediated immunity** involves the production of special proteins called antibodies by B-cells, which circulate around the body and attack invading agents.
- The **cell-mediated response** is due to T-cells and involves the formation of special lymphocytes that destroy invading agents.

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Antigens

Antibody-mediated and cell-mediated immunity are both triggered by antigens. An **antigen** is any substance capable of causing a specific immune response. They are large molecules such as proteins, carbohydrates, lipids or nucleic acids and may include (among others):

- virus particles
- whole micro-organisms, such as a bacterial cell
- part of a bacterium, such as the flagella, cell wall or capsule
- toxins
- molecules on cells such as blood cells
- pollen grains
- egg whites.

Large molecules produced in a person's own body do not cause an immune response. These are called **self-antigens**. Foreign compounds that do trigger an immune response are **non-self antigens**. The immune system becomes programmed before birth to distinguish between self-antigens and non-self antigens. From then on, it only attacks non-self antigens.

Key concept

Antigens are capable of producing an immune response.

Antibodies

An **antibody** is a Y-shaped specialised protein that is produced by **plasma cells** in response to a non-self antigen. Antibodies belong to a group of proteins known as **immunoglobulins**, often represented as 'Ig'. There are five classes of antibodies, which vary in their structure and are designated IgA, IgD, IgE, IgG and IgM.

The antibody produced in response to an antigen can combine with that antigen to form an **antigen–antibody complex**. Antigen molecules have specific active sites with a particular shape. The antibody has the complementary shape, allowing the two molecules to fit together like a key in a lock. Each antibody can combine with only one particular antigen, in the same way that a key will only open a particular lock.

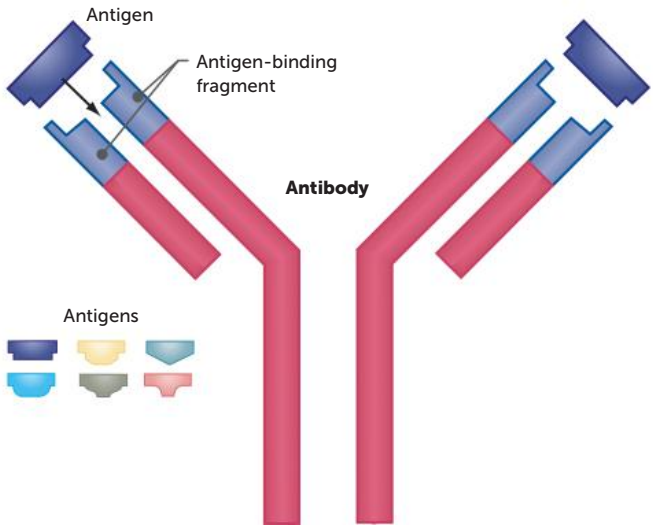

FIGURE 7.18
The active sites on the antibody and antigen molecules are complementary: they fit together like a lock and key

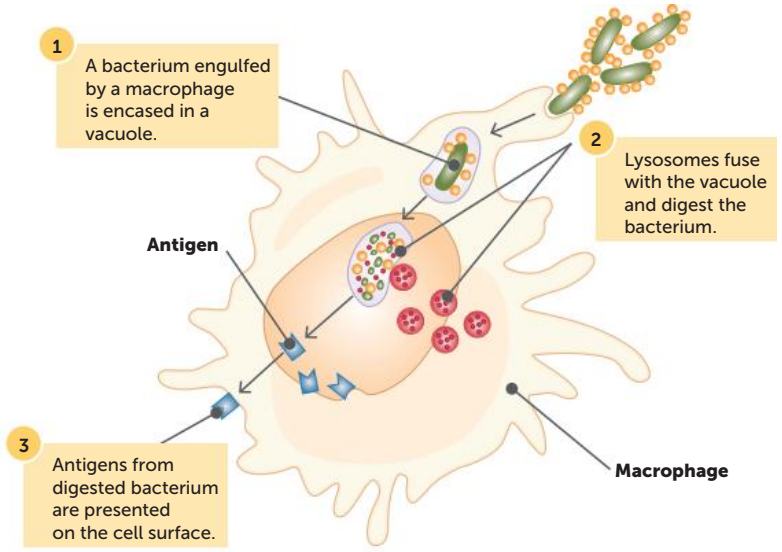

FIGURE 7.19 Antigen-presenting cells, such as macrophages, detect, engulf and digest pathogens to present the antigens to lymphocytes.

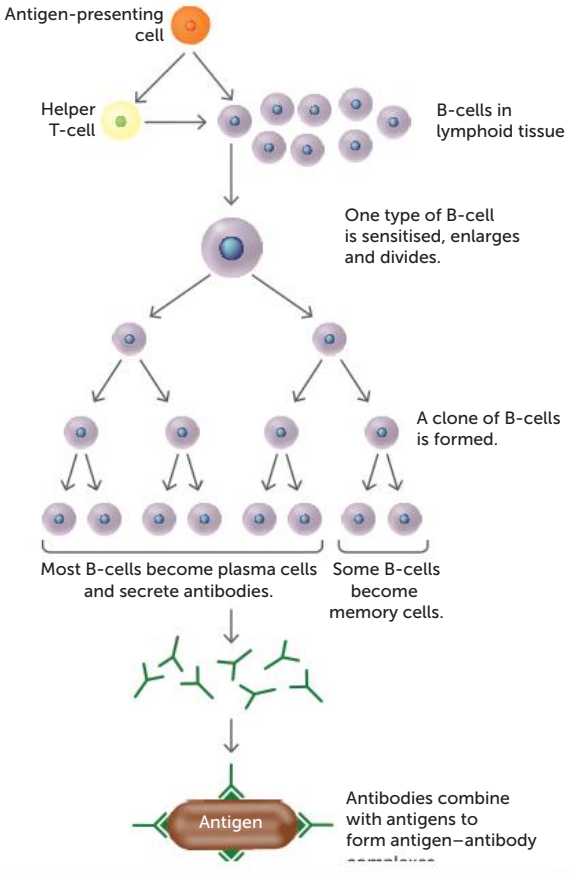

FIGURE 7.20 Events in the antibody-mediated immune response

Antigen-presenting cells

When a non-self antigen enters a body, specific cells recognise this and respond appropriately. These cells are called **antigen-presenting cells**, and include dendritic cells, macrophages and undifferentiated B-cells. These cells:

- detect the presence of a non-self antigen
- engulf the pathogen
- digest the pathogen, producing small fragments that move to the surface of the cell
- present the antigen to lymphocytes.

Antibody-mediated immunity

The humoral response involves the production and release of antibodies into the blood and lymph. This is antibody-mediated immunity. It provides resistance to viruses, bacteria and bacterial toxins before these micro-organisms or substances enter the body's cells.

Lymphoid tissue contains thousands of types of B-cells. Each type has a receptor for a particular antigen and, therefore, is capable of responding to a specific antigen. When an antigen-presenting cell presents the antigen to the specific B-cells, the B-cells are activated. The antigen is also present to helper T-cells, leading to the release of **cytokines**, small proteins that are released in response to antigens and act as messengers in the immune response. These cytokines cause the helper T-cells to clone themselves to release different cytokines, which activate the B-cells.

When the B-cells are activated, they enlarge and divide into a group of cells called a **clone**. Most of the clone becomes plasma cells, which secrete the specific antibody capable of attaching to the active site of the antigen. These antibodies circulate in the blood, lymph and extracellular fluid to reach the site of the invasion of micro-organisms or foreign material. The remaining B-cells of the clone become **memory cells**. Memory cells spread to all body tissues to allow the response to occur more rapidly should the antigen enter the body again.

On the first exposure to an antigen the immune reaction is called the **primary response**. The body's immune system usually responds fairly slowly, often taking several days to build up large amounts of antibodies. This is because it takes time for the B-cells to multiply and differentiate into plasma cells and then secrete antibodies. Once the level of antibodies reaches a peak, it begins to decline. However, the primary response leaves the immune system with a memory of that particular antigen.

With a second or subsequent exposure to the same antigen, the response is much faster due to memory cells recognising the antigen more quickly. With this **secondary response**, plasma cells are able to form very quickly, with antibody levels in the blood plasma rising rapidly to a higher level that lasts longer. Frequently, this response is so quick that the antigen has little opportunity to exert any noticeable effect on the body and no illness results.

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How antibodies work

Different antibodies protect the body by different methods. They may:

- inactivate foreign enzymes or toxins by combining with them or inhibiting their reaction with other cells or compounds
- bind to the surface of viruses and prevent them entering cells
- coat bacteria so that they are more easily consumed by phagocytes
- cause particles such as bacteria, viruses or foreign blood cells to clump together, a process known as **agglutination**
- dissolve organisms
- react with soluble substances to make them insoluble and thus more easily consumed by phagocytes.

Figure 7.24 summarises the action of antibodies.

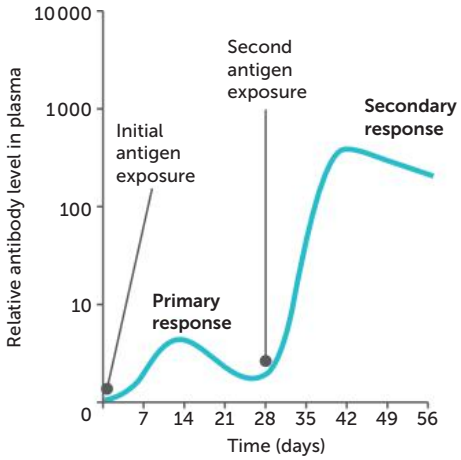
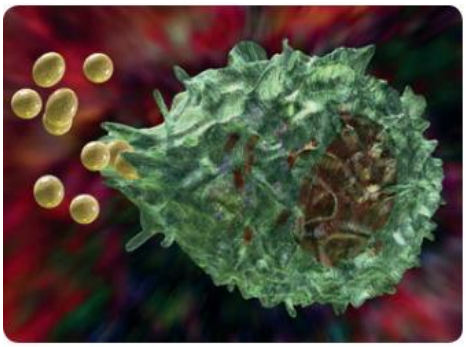


FIGURE 7.21 The antibody level in blood plasma after a first and second exposure to an antigen



Science Photo Library/Russell Kightley

FIGURE 7.22 A macrophage (green) engulfing bacteria (yellow) by the process of phagocytosis

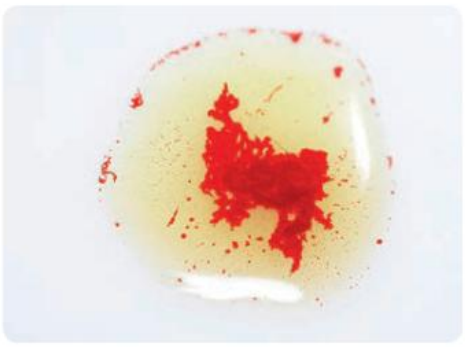


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FIGURE 7.23 Agglutination of red blood cells due to the presence of antibodies for the antigens on the surface of the cells

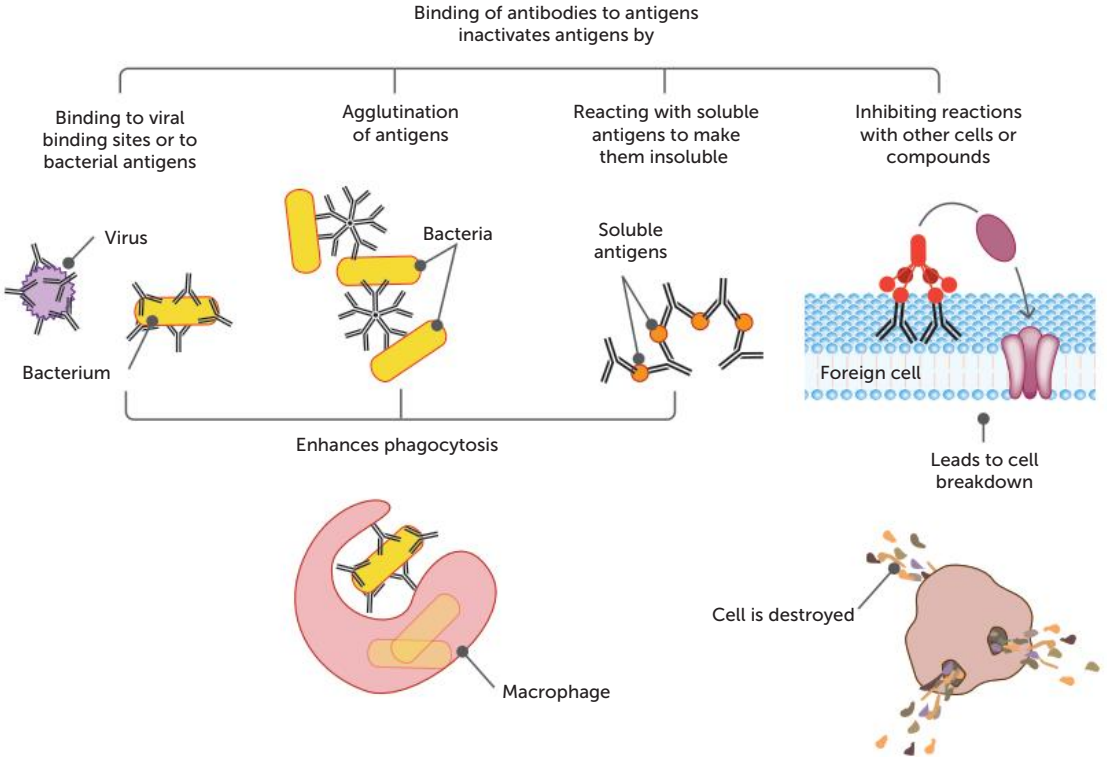

FIGURE 7.24 Summary of how antibodies interact with antigens to inactivate the antigens

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Immune response
This website provides an animated sequence showing the immune response.

Key concept

Antibody-mediated immunity occurs when B-cells are stimulated, resulting in the production of antibodies and memory cells.

Cell-mediated immunity

Cell-mediated immunity provides resistance to the intracellular phase of bacterial and viral infections. These pathogens, such as the bacteria responsible for tuberculosis and Legionnaire's disease, specialise in invading and replicating inside their hosts' own cells, making them particularly difficult to overcome.

Cell-mediated immunity is also important in 'fighting' whole cells, such as providing resistance to fungi and parasites, and in rejecting foreign-tissue transplants. It also appears to be important in fighting cancer cells.

The T-lymphocytes are responsible for cellular immunity. They occur in the same lymphoid tissue as B-cells but occupy different areas of the tissue. Like B-cells, there are thousands of types of T-cells, and each type responds only to one particular antigen. When a foreign antigen such as a virus or a bacterium enters the body, the antigen-presenting cells present the antigen to the particular type of T-cells. These become activated or sensitised.

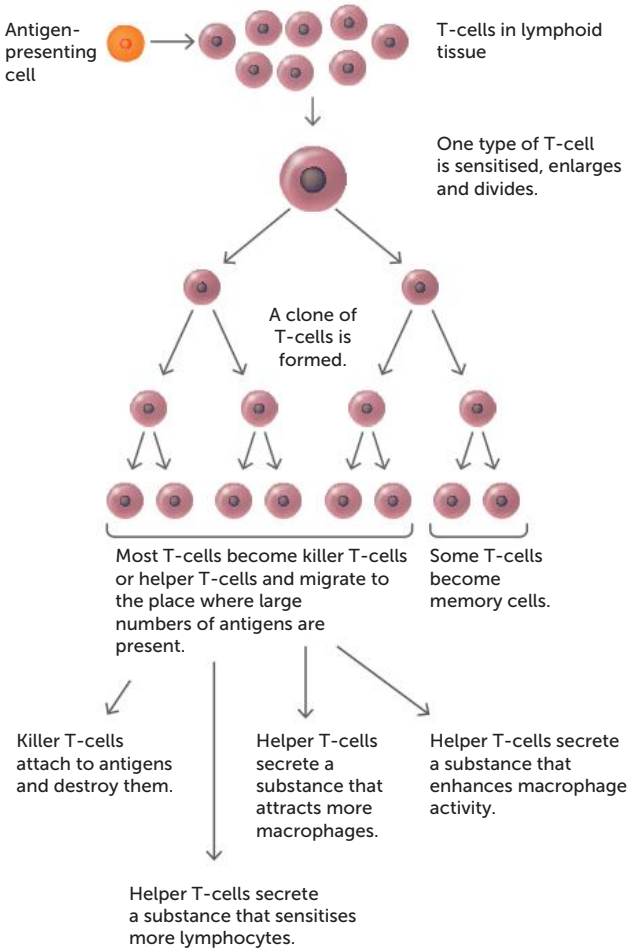

FIGURE 7.25 Response to T-cells in cell-mediated immunity

The sensitised T-cells enlarge and divide, each giving rise to a clone, a group of identical T-cells. Some cells of the clone remain in the lymphoid tissue as memory cells, which are able to quickly recognise the original invading antigen. If infection with the same antigen should occur again, these memory cells can initiate a much faster response to the second and subsequent infections.

The T-cells that do not become memory cells develop further, producing three different types of T-cell.

1. **Killer T-cells** (also known as **cytotoxic T-cells**) migrate to the site of infection and deal with the invading antigen. They attach to the invading cells and secrete a chemical that will destroy the antigen, and then go in search of more antigens.
2. **Helper T-cells** play an important role in both humoral and cellular immunity. They bind to the antigen on antigen-presenting cells, stimulating the secretion of cytokines that:
   - attract lymphocytes to the infection site which become sensitised and activated, thus intensifying the response

- attract macrophages to the place of infection so that the macrophages can destroy the antigens by phagocytosis
- intensify the phagocytic activity of macrophages
- promote the action of killer T-cells.

3 **Suppressor T-cells** act when the immune activity becomes excessive or the infection has been dealt with successfully. They release substances that inhibit T- and B-cell activity, slowing down the immune response.

Key concept

Cell-mediated immunity occurs when T-cells are stimulated, resulting in the production of killer T-cells and helper T-cells as well as memory cells.

TABLE 7.2 Summary of immune responses

ANTIBODY-MEDIATED IMMUNITY (HUMORAL IMMUNITY)	CELL-MEDIATED IMMUNITY (CELLULAR IMMUNITY)
Works against bacteria, toxins and viruses before they enter the body's cells; also against red blood cells of a different blood group than the person. 1 Antigen-presenting cells recognise, engulf and digest pathogens, displaying the antigen on their surface. 2 Antigen-presenting cells reach lymphoid tissue and present the antigen to lymphocytes. 3 Helper T-cells are stimulated by antigen-presenting cells, which release cytokines. 4 Specific B-lymphocytes are stimulated to undergo rapid cell division. 5 Most new B-cells develop into plasma cells, which produce antibodies and release them into blood and lymph. 6 Antibodies combine with the specific antigen and inactivate or destroy it. 7 Some of the new B-cells form memory cells.	*Works against transplanted tissues and organs, cancer cells and cells that have been infected by viruses or bacteria; also provides resistance to fungi and parasites.* 1 Antigen-presenting cells recognise, engulf and digest pathogens, displaying the antigen on their surface. 2 Antigen-presenting cells reach lymphoid tissue and present the antigen to the lymphocyte. 3 Helper T-cells are stimulated by antigen-presenting cells, which release cytokines. 4 Specific T-lymphocytes are stimulated to undergo rapid cell division. 5 Most new T-cells develop into killer T-cells or helper T-cells, which migrate to the site of the infection. 6 Killer T-cells destroy the antigen, while helper T-cells promote phagocytosis by macrophages. 7 Some sensitised T-cells form memory cells.

Immune response
This website includes an animation of a B-cell and a T-cell attacking an invader.

Types of immunity

Immunity is resistance to infection by invading micro-organisms. The presence of memory cells allows the body to respond quickly enough to deal with any invasion by pathogenic micro-organisms before symptoms of disease occur. The ability to respond rapidly may be natural or artificial. **Natural immunity** occurs without any human intervention; **artificial immunity** results from giving people an antibody or antigen.

Natural and artificial immunity can be passive or active.

Passive immunity

Passive immunity is when a person receives antibodies produced by someone else, meaning that the individual's body plays no part in the production of antibodies. This can occur naturally when antibodies from the mother pass across the placenta to a developing foetus or when the mother's antibodies are passed to the baby in breast milk. It can also be gained artificially when a person is injected with antibodies to combat a particular infection. This is often done when a person is exposed to pathogens that cause serious diseases, such as tetanus, diphtheria and rabies. Antibodies are given so that immunity is established immediately. Passive immunity is short-lived: it lasts only until the antibodies are broken down and excreted.

Active immunity

Active immunity results when the body is exposed to a foreign antigen and manufactures antibodies in response to that antigen. While the amount of antibody decreases, this type of immunity lasts longer than passive immunity due to the presence of memory cells. Should a subsequent infection involving the same antigen occur, the appropriate antibodies can be produced very quickly, eliminating the antigen before the infection can produce any disease symptoms. Active immunity to a disease can develop from having the disease and recovering (natural active immunity) or from an injection of the antigens associated with the disease (artificial active immunity).

Table 7.3 summarises the types of immunity.

TABLE 7.3 Types of immunity

	NATURAL	ARTIFICIAL
Passive	Antibodies enter the bloodstream across the placenta or in breast milk.	Antibodies are injected into the bloodstream.
Active	Ability to manufacture antibodies results from an attack of a disease.	Ability to manufacture antibodies results from being given an antigen by vaccination.

Key concept

Immunity is due to memory cells that react very quickly when exposed to the specific antigen. It may be passive or active, and be gained naturally or artificially.

Questions 7.3

RECALL KNOWLEDGE

1. Define 'specific defences'.
2. Name the two types of lymphocytes, and state where each is produced and becomes mature.
3. Which type of lymphocyte is responsible for cell-mediated immunity?
4. Define 'antigen' and describe its role in specific defences.
5. Describe the structure of an antibody.
6. Describe the series of events that occur during antibody-mediated immunity.
7. List the ways that an antigen–antibody complex stops an infection.
8. Describe the function of helper T-cells, killer T-cells and suppressor T-cells.
9. Describe how immunity can be classified based on the method of gaining:
   a. antibodies
   b. immunity.

APPLY KNOWLEDGE

10. Draw a flow chart to show how the immune response is a homeostatic response.
11. People whose blood type is A have the A antigen on their red blood cells.
    a. Explain why they may contain B antibodies in the plasma.
    b. Suggest what might happen if they are given red bloods cells of blood type B or AB.
    c. Explain why red blood cells of blood type O are safe to receive.
    d. Explain why, if a plasma transfer is needed, the preferred type is A and not O.
12. Why is the secondary response quicker and longer lasting than the primary response?
13. Even though we consider the cell-mediated and humoral responses separately, there is some overlap. Discuss this overlap.
14. For each situation below, state whether the humoral response or cell-mediated response would be more important.
    a. A heart transplant
    b. A viral infection
    c. A blood transfusion
    d. Tetanus toxins
    e. A bacterial skin infection
    f. A fungal infection.
15. Explain why vaccination leads to an active, artificial immunity, while breast milk produces a passive, natural immunity in a baby.

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7.4 PREVENTION AND TREATMENT OF DISEASE

Understanding disease, and our body's response to it, allows us to develop methods of preventing and treating it.

Vaccines

Immunisation means programming the immune system so that the body can respond rapidly to infecting micro-organisms. In other words, it is developing an immunity. This can occur naturally or artificially. **Vaccination** is the artificial introduction of antigens of pathogenic organisms so that the ability to produce the appropriate antibodies is acquired without the person having to suffer the disease. Thus, there is a slight difference in meaning between 'vaccination' and 'immunisation', but the two words tend to be used interchangeably.

A **vaccine** is the antigen preparation used in artificial immunisation. Traditional vaccines are of four types.

- *Live attenuated vaccines*: Living **attenuated** micro-organisms are micro-organisms of reduced **virulence**; that is, micro-organisms with a reduced ability to produce disease symptoms. Therefore, the immunised person does not contract the disease but manufactures antibodies against the antigen. Vaccines containing living attenuated micro-organisms include those for immunisation against polio, tuberculosis, rubella (German measles), measles, mumps and yellow fever.

Science Photo Library/James King-Holmes

FIGURE 7.26 Vaccines for some viral diseases are produced by allowing the viruses to multiply in living cells: here the influenza virus is being introduced into fertile eggs

TABLE 7.4 Deaths from diseases commonly vaccinated against, Australia, 1926–2016

PERIOD	DIPTHERIA	PERTUSSIS	TETANUS	POLIOMYELITIS	MEASLES	POPULATION ESTIMATE (MILLION)
1926–1935	4073	2808	879	430	1102	6.60
1936–1945	2791	1693	655	540	822	7.20
1946–1955	624	429	625	1091	495	8.60
1956–1965	44	58	280	176	210	11.00
1966–1975	11	22	82	61	146	13.75
1976–1985	2	14	31	70	62	14.90
1986–1995	2	9	21	69	36	17.30
1996–2005	0	9	7	140	1	18.73
2006-2016	3	36	7	183	2	20.80

Source: AIHW: Vaccine-preventable diseases fact sheets, data tables, November 2018. Australian Institute of Health and Welfare (AIHW) CC-BY 3.0.

Note 1: Shading indicates decade in which community vaccination started for the disease.

Note 2: Since the widespread introduction of the polio vaccine in the 1950s, death caused by polio has been rare. The majority (89%) of polio deaths after 1996 were in people aged 65 and older and were likely due to the after-effects of a previous infection.

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- *Inactivated vaccines*: Inactivated vaccines contain dead micro-organisms. They produce an immunity that is shorter lasting than immunisation using live attenuated micro-organisms. Examples of vaccines of this type include cholera, typhoid and whooping cough vaccines.
- *Toxoid vaccines*: In cases where bacteria produce their effects in humans by liberating toxins, it is not necessary to use the bacteria for immunisation. The toxins produced by the bacteria can be inactivated, so that when they are injected into someone they do not make the person ill. Such inactivated toxins are called **toxoids**. Injections of toxoids are used to immunise against diphtheria and tetanus.
- *Sub-unit vaccine*: Instead of using a whole dead or attenuated micro-organism, a fragment of the organism can be used to provoke the immune response. Sub-unit vaccines are used for vaccination against human papilloma virus (Gardasil) and hepatitis B.

Pasteur
This website provides more information on the work of Louis Pasteur and the discovery of vaccines.

Scientists are constantly investigating alternative methods of immunisation that are more effective with fewer side effects. One approach is to modify the characteristics of the pathogen by slightly changing the DNA in the micro-organism's cell, making the pathogen less virulent. Another method is to insert certain DNA sequences from the pathogen into harmless bacterial cells. The chosen DNA sequence causes the production of antigens that are characteristic of the pathogen. Vaccination with the harmless bacterium results in immunity against the pathogen. It is likely that a great many future vaccines will be made using this **recombinant DNA** method. Recombinant DNA will be discussed further in Chapter 8.

Vaccine delivery

The most common method of vaccination is to inject the vaccine using a syringe, but other forms of delivery can be used. One type of polio vaccine is given by mouth in a sweet syrup or in lumps of sugar. This method is no longer used in Australia but is still in use in many countries. Other forms of delivery are currently under research, including a fine spray, skin patches and ingestion in food.

Dreamstime.com/Paulus Rusyanto

FIGURE 7.27 Childhood vaccinations greatly reduce certain illnesses in children and also prevent the spread of communicable disease

Vaccination schedule

Most vaccinations do not start until a child is two months old, and for most diseases more than one vaccination is necessary.

Vaccination should not start too soon after birth, as the child's blood contains antibodies from its mother via the placenta or in breast milk. If a newborn is given a vaccine, the antibodies from the mother eliminate the antigens in the vaccine. This occurs before the child's immune system can mount an immune response. A few months are also necessary for the child's immune system to become activated and therefore able to prevent the child from getting the diseases that they are being vaccinated against. The hepatitis B vaccine is an exception to this, due to the risk of the infant being infected during birth. Therefore, the first vaccine is given soon after birth to provide early protection for the baby.

Unfortunately, one injection of a vaccine is not usually enough to protect a person from the particular disease they are being vaccinated against. The antibody levels from the primary response following the first vaccination will decline. Therefore, a second vaccination, called a booster, is needed stimulate a secondary response. The memory cells react quickly to this second exposure, resulting in a higher, longer-lasting level of antibodies in addition to more memory cells.

The timing of a booster shot is important. If the booster is given too soon after the first vaccination, the antibodies present in the blood will eliminate the material in the vaccine before more B-cells can be activated. To avoid this, a period of time between vaccinations is required, to allow the antibodies in the blood to be eliminated. Usually, this takes around two months.

In Australia, most people are vaccinated against the diseases for which vaccines are available. Table 7.5 shows a recommended vaccination schedule for Australians from birth through to adult life. For Australians travelling overseas, other vaccinations such as cholera, yellow fever and typhoid may be recommended, depending on the destination.

Key concept

Vaccines work by stimulating the immune system to promote immunity for an antigen. More than one vaccination is often needed to develop sufficient levels of antibodies and memory cells to protect the individual.

TABLE 7.5 Recommended vaccination schedule for Australians

AGE	RECOMMENDED VACCINATION
Birth	Hepatitis B
2 and 4 months	Diphtheria, tetanus, whooping cough; polio; hepatitis B; *Haemophilus influenzae* type B (HiB); rotavirus*; pneumococcal**
6 months	Diphtheria, tetanus, whooping cough, polio, HiB, rotavirus, pneumococcal, hepatitis B
12 months	Measles, mumps, rubella (MMR), HiB, meningococcal C***
18 months	Measles, mumps, rubella, chickenpox
4 years	Diphtheria, tetanus, whooping cough, polio, measles, mumps, rubella (MMR only if not given at 18 months)
10–15 years	Hepatitis B, chickenpox, diphtheria, tetanus, whooping cough, human papillomavirus (HPV)
15 years and over	Influenza (for Aboriginal and Torres Strait Islander people), pneumococcal (for Aboriginal and Torres Strait Islander people medically at risk)
50 years and over	Influenza (for Aboriginal and Torres Strait Islander people), pneumococcal (for Aboriginal and Torres Strait Islander people)
Pregnant women	Influenza
65 years and over	Influenza (annually), pneumococcal

Source: NHMRC Australian Standard Vaccination Schedule, 1 July 2013.

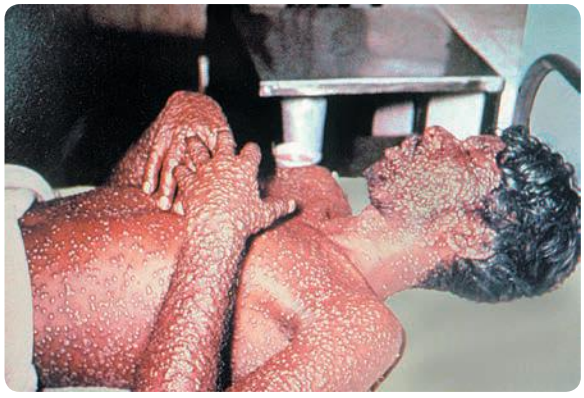
Notes: *Protects against a highly infectious disease of the small intestine; most cases occur in children under five years.

**Protects against a bacterial infection of the lung that may lead to pneumonia if it occurs in children or the elderly.

***Protects against a bacterial infection of the membranes around the brain.

Vaccination of populations

The World Health Organization (WHO) rates the introduction of vaccines as one of the public health measures that has had the greatest impact on people's health. The use of vaccines in mass immunisation programs has either eradicated or greatly reduced the incidence of certain diseases throughout the world. WHO's greatest success is probably the global elimination of smallpox. The last known naturally occurring case was in Somalia in 1977, although a small number of laboratory-acquired infections have occurred since then. The WHO is now determined to eliminate polio using a range of vaccination programs. Since 1988, the number of polio cases globally has decreased from about 350 000 a year to 22 cases in 2017.



Alamy Stock Photo/Science History Images

FIGURE 7.28 Smallpox causes a rash with raised pustules filled with pus-like fluid

Other vaccination programs are on a smaller scale and are frequently used to prevent the possibility of a serious outbreak of a highly infectious disease. In Australia, prior to winter each year, the federal government supports a program to vaccinate the young and the elderly against current strains of the influenza virus. Such vaccination programs not only reduce the chance of disease in the most susceptible individuals but also increase the immunity of the population. Such immunity is referred to as **herd immunity** and depends on a high proportion of individuals being immunised. When there are a large number of immune individuals in a population, there is less chance of the disease being transmitted between them.

The proportion of the population that needs to be immune to protect the population varies between diseases. Highly contagious diseases, such as measles, need a very high percentage of the population to be vaccinated to provide protection. However, a smaller percentage is needed for protection from less contagious diseases, such as Ebola.

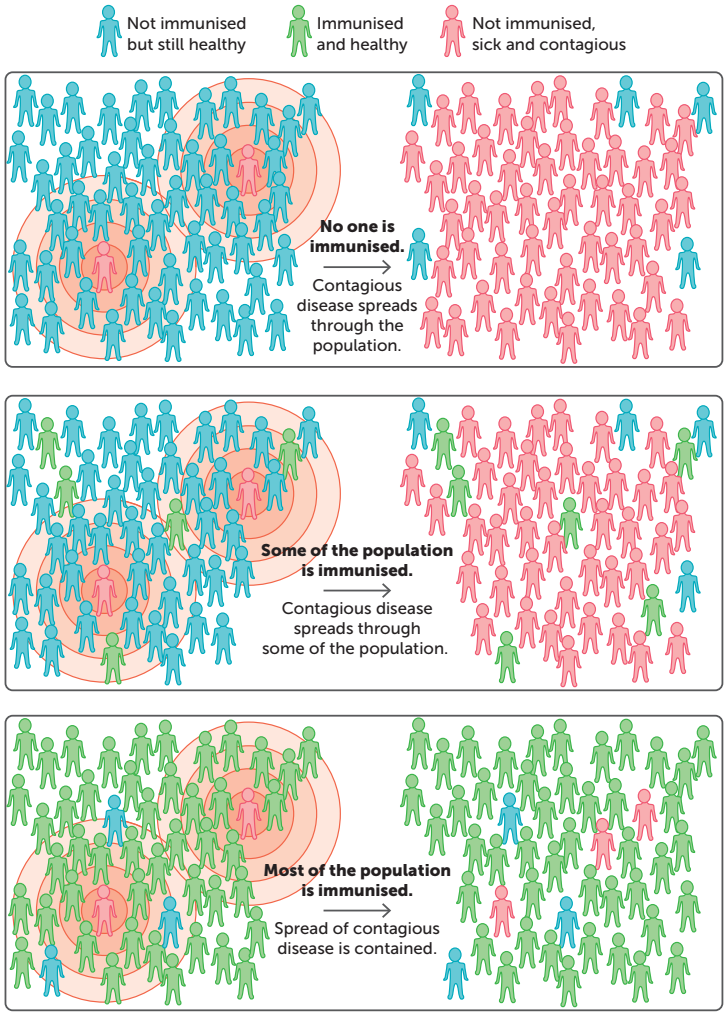


FIGURE 7.29 Herd immunity protects the whole population

Herd immunity
This website contains more information about herd immunity

Herd immunity simulation
This website has a simulation comparing the spread of disease in communities with different levels of immunisation.

One problem for health departments in all countries is that, as the incidence of infectious diseases declines, people become complacent and may decide that the risk of side effects from the vaccine is higher than the risk of contracting the disease itself. If vaccination rates do decline, a serious outbreak of a disease may occur. This happened in the United Kingdom in the 1970s, resulting in large outbreaks of whooping cough. Even in countries where vaccination rates are high, vaccine-preventable diseases have sometimes reappeared. The Netherlands, for example, has one of the highest rates of fully vaccinated people in the world. Nevertheless, there are groups of Dutch people who object to vaccination on religious grounds. In the early 1990s, a large outbreak of polio affected these people, with some suffering severe complications such as paralysis. However, polio did not spread into the rest of the Dutch community because of the protection provided by the high rates of vaccination.

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Key concept

Herd immunity relies on a large proportion of the population being immune to a disease to protect the whole population.

Factors to consider with vaccinations

One of the important choices that parents must make is whether to have their children vaccinated in infancy. Childhood vaccination is not compulsory in Australia, but in 2019 the Australian Government Department of Health reported that 94.31% of infants had been vaccinated by the age of 12 months.

There are many reasons to get vaccinated. Vaccinations provide protection for both individuals and whole populations. Negative side effects, while possible, are rare. Additionally, there are often reduced costs for things like health care, and in some states, such as Western Australia, it is a requirement of enrolment in a child-care or educational establishment.

As with all medical procedures, there are risks involved in the use of vaccines. However, there are strict guidelines that aim to minimise the possible risks. Before vaccines are made available for general use, they are tested for safety and effectiveness, first in clinical trials and then in much larger trials. In Australia, all vaccines on the market are manufactured according to strict safety guidelines. Before marketing approval is granted, they are evaluated by the Therapeutic Goods Administration to ensure they are effective and are at a high standard of quality and safety.

In Australian society, the Internet and other media are major sources of misinformation about the risks and benefits of immunisation. In 2014, the NSW Healthcare Complaints Commission ordered an anti-vaccination website to change its name, and a warning was issued about the unreliability of information published on that site. When accessing the Internet for material about immunisation, it is essential to check the reliability of the sources.

Various factors may affect a person's viewpoint on vaccines.

Inability to be vaccinated due to health issues

- *Allergic reactions*: One of the main risks of vaccination is an allergic reaction. This may occur, not so much from the vaccine itself, but from a reaction to the medium in which the vaccine was cultured. The National Health and Medical Research Council lists the possible vaccine components that may result in an allergic response. For example, many of the influenza vaccines are manufactured in fertilised eggs, and people who are allergic to egg protein need to be aware of this. Similarly, people who are allergic to yeast would need to be mindful that some of the older hepatitis B vaccines have yeast as a component.
- *Preservatives*: In the manufacture of vaccines, certain chemicals are used as preservatives. Preservatives used include thiomersal, formaldehyde, phenol (carbolic acid), aluminium phosphate, alum and acetone. Individuals concerned about vaccination claim that these preservatives can affect the nervous system and lead to other health issues. Such claims have been investigated on a number of occasions, and no connection has been identified. Instead, it appears that any reaction is due to chance alone.

Vaccination safety
This article from the *Medical Journal of Australia* discusses the risks associated with vaccines.

Social factors

- *Ethical concerns with the use of animals to produce vaccines*: As viruses can only reproduce in living cells, the manufacture of viral vaccines requires host tissue. For example, influenza virus is cultured in chicken embryos and Japanese encephalitis virus is grown in the brains of mice. Consequently, some people are concerned about the treatment of animals in the production of vaccines.
- *Ethical concerns with the use of human tissue to produce vaccines*: Many vaccines require human tissue because some viruses that cause disease in humans do not grow well in cells derived from other species. In addition, the use of human tissue avoids the problems of cross-species infection from possible unknown viruses. The source of the human tissue is a concern for many people. For example, rubella vaccine is manufactured using cultured human

cells. The original cells for the cultures were obtained from human foetuses. This raises moral questions for people who are opposed to the way in which those original cells were obtained.

- *Ethical concerns with informed consent*: A key principle of ethics is informed consent, and this applies to trialling vaccines. There is some concern that trailling vaccines in developing countries may lead to their use in populations with low standards of education. This may mean that people are not fully aware of the risks and may be open to exploitation by the vaccine's manufacturer.
- *Ethical concerns with testing on animals*: Before clinical trials on humans, most vaccines are tested on animals to identify problems that could arise in humans. The animals used in such testing are frequently mice, but other mammals are also used, along with birds, amphibians and fish. Legislation exists to limit the way that animals can be used; however, some people do not believe they should be used at all.
- *Concerns about promoting sexual activity in teenagers*: Some people believe that vaccinating against the sexually transmitted infection human papilloma virus will likely encourage teenagers to be sexually active.
- *Availability*: Vaccines may not be readily available in all areas.

Cultural factors

- *Religious beliefs*: Religious belief has often been cited as a reason for some Australian parents refusing to immunise their children. However, none of the major religions in Australia – Christianity, Islam and Judaism – are opposed to immunisation. There are, however, a few religions that are opposed to vaccines. These are religions that rely on faith healing or healing through prayer, such as Church of the First Born and First Church of Christ. In addition, the methods used to produce vaccines may contradict religious beliefs and lead to a choice not to participate in immunisation programs.

Religion and vaccines
This article discusses the religions that are opposed to vaccines.

Economic factors

- *Cost of vaccine*: The vaccines may be too expensive for some people to afford.
- *Commercialisation*: The interests of commercial vaccine production may affect its use.

In all issues relating to vaccines, individuals must be guided by their own beliefs and values. However, before making a decision about vaccination, it is the responsibility of each of us to ensure we are fully informed about all the possible consequences of the decision.

7.2 Specific resistance to infection

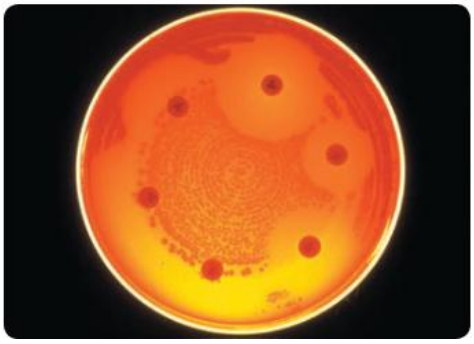
Activity 7.4
Investigating the testing of animals in the manufacture of vaccines

Key concept

The decision to participate in immunisation programs should be made after careful consideration of the risks and benefits, as well as any social, cultural or economic considerations.

Antibiotics

Antibiotics are drugs that are used to fight infections of micro-organisms, particularly bacteria. They cannot be used to treat viral infections. Each antibiotic is effective for only certain types of bacterial infection, and testing is often carried out prior to antibiotics being prescribed.



Medical Images RM/Bob Tapper

FIGURE 7.30
Comparing antibiotic sensitivity of bacteria using a culture plate with antibiotic assay discs. The discs surrounded by a clear area contain antibiotics that are effective for this type of bacteria

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Before antibiotics came to be widely used in the 1940s, a person could die from an infected cut or scratch that today would be considered a minor problem. The discovery of antibiotics brought about a revolution in the treatment of bacterial infections, and they are now one of the most frequently prescribed drugs.

The first antibiotic to be identified was penicillin, when it was discovered that the mould *Penicillium notatum* was able to stop the growth of the *Staphylococcus* bacteria. Penicillin works by preventing the synthesis of the walls of the bacterial cells, inhibiting the reproduction of bacteria. About 30% of antibiotics used in Australia today are penicillin based, including amoxicillin and ampicillin. However, the effectiveness of penicillin has been reduced because many bacteria have developed resistance to it. Also, about 10% of people are allergic to penicillin.

Since the discovery of penicillin, a number of different antibiotics have also been developed, including:

- Streptomycin, erythromycin, neomycin, tetracycline and vancomycin, which interfere with protein synthesis in the cells of the target bacteria.
- Cephalosporin, which interferes with synthesis of the cell wall. It is much less likely than penicillin to produce allergic reactions.

There are two types of antibiotics. **Bactericidal antibiotics** kill bacteria by changing the structure of the cell wall or cell membrane, or by disrupting the action of essential enzymes. **Bacteriostatic antibiotics** stop bacteria from reproducing, usually by disrupting protein synthesis. Both types are effective in treating bacterial infections.

Some antibiotics affect a wide range of different types of bacteria. These are **broad-spectrum antibiotics**. Others, **narrow-spectrum antibiotics**, are effective only against specific types of bacteria.

The widespread use of antibiotics has created a major problem. Some of the bacteria that antibiotics are used to kill have gradually evolved and become resistant to them. In the early days of antibiotic use, the problem was easily solved by changing to a different antibiotic. However, some strains of bacteria are now resistant to most or all available types of antibiotics. This is known as **multiple drug resistance** and such bacteria are often referred to as 'super bugs'. In 2012 it was reported that 12 cases of tuberculosis in Mumbai, India, were resistant to all known drugs; that is, they showed **total drug resistance**. Totally resistant strains of the bacterium that causes gonorrhoea have been detected in Australia, Japan and Europe.

Multiple drug resistance has been hastened by the overuse of antibiotics in medicine and in agriculture. Doctors have prescribed antibiotics to prevent infection rather than to treat an existing infection. Farmers use antibiotics as 'growth promoters' in poultry, pigs and cattle. International efforts are now being made to reduce the use of antibiotics so that the development of further strains of multiple drug-resistant bacteria will be delayed.

Prevention of misuse and abuse of antibiotics will slow the development of resistance, but there is no way of stopping it altogether. Strategies being used to overcome the problem are to develop new classes of antibiotics to which bacteria have no resistance, to revive old antibiotics by using them in combination with other substances, and to genetically engineer bacteria to disable antibiotic-resistant genes.

Discovery and development of penicillin
This website looks at the discovery of penicillin.

Antibiotics
This website provides information on how antibiotics work.

Activity 7.5
Investigating antibiotic resistance

Antivirals

Antiviral drugs are used specifically for treating viral infections. Because antibiotics are ineffective against viruses, there is still no treatment for common ailments such as colds, chickenpox and measles. This has led to a hunt for chemicals that can be used as antivirals.

Viruses enter a host cell, and the virus DNA or RNA induces the cell to produce new virus particles. These particles can then leave the cell and infect new host cells. The way in which viruses replicate makes it difficult to find drugs that will treat viral infections. Because the host cell produces the new virus particles, any drug that interferes with virus replication is likely to be toxic to the host.

Early research involved culturing cells, infecting them with a virus and then trying different chemicals to see whether the amount of virus decreased. This time-consuming and hit-or-miss technique produced little result.

In the 1980s, it became possible to determine the genetic sequences of viruses so that scientists could find out exactly how they work. Research today is aimed at identifying viral proteins that can be disabled by specially designed chemicals. If the proteins are very different from human proteins, there should be few side effects from the use of such drugs.

Unlike most of the antibiotics that destroy pathogenic bacteria, antivirals inhibit the development of the virus. Most of the antiviral drugs that are now available target HIV, herpes, hepatitis B and C, and influenza A and B.

Some examples of antivirals that you may have heard of are Tamiflu and Relenza for influenza, acyclovir (marketed as Zovirax) for herpes infections, interferons for hepatitis B and C, and zidovudine (AZT) for human immunodeficiency virus. A great deal of research is being carried out to develop drugs that will target other viruses such as coronavirus.

Key concept

Antibiotics are used to treat bacterial infections, and antiviral drugs are used to treat viral infections.

Questions 7.4

RECALL KNOWLEDGE

1. Define 'vaccination'.
2. List the ways that vaccines have traditionally been made.
3. Why are infants not vaccinated against most diseases until at least two months of age?
4. Define 'herd immunity' and describe what is needed to achieve it.
5. List some of the reasons that people may choose not to be vaccinated.
6. Penicillin is an antibiotic.
   - a Define 'antibiotic'.
   - b List two other antibiotics.
   - c Would penicillin be effective in treating an infection caused by the influenza virus? Explain.
7. Define 'antiviral drug' and identify an infection that is able to be treated with antiviral drugs.

APPLY KNOWLEDGE

8. A person whose immune system is compromised is unable to be vaccinated. Explain why this is so, and why herd immunity plays a vital role in this person's health.
9. Explain how it is possible to introduce a virus or bacteria in a vaccine without producing the associated disease.
10. Which antibiotic is more important – broad-spectrum or narrow-spectrum? Justify your answer.
11. 'Golden staph' is a common name for the bacteria *Staphylococcus aureus* that can be resistant to most commonly used antibiotics. Explain why this bacteria is such a problem.
12. Explain why antiviral drugs are harder to develop than antibiotics.
13. Use a table to compare and contrast bacteria and viruses in terms of:
    - a their size
    - b their structure
    - c whether they are living or non-living
    - d how they replicate
    - e how they affect the body
    - f treatment.

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CHAPTER 7 ACTIVITIES

ACTIVITY 7.1 Investigating the effectiveness of hand washing

Hand washing is recommended as a way of reducing the spread of bacteria. Antiseptics are used to further reduce the risk of infection.

The purpose of this activity is to compare the effectiveness of different methods of hand washing or types of soap.

The presence of bacteria on the skin of the fingers can be demonstrated by pressing the fingers on to the surface of the medium in a sterile culture plate, a petri dish with a thin layer of agar jelly in the bottom. After pressing the fingers on to the agar, the plate is incubated (kept in a warm place) for several days. Bacteria that were transferred from the fingers to the agar will reproduce and form colonies that can be seen with the naked eye. The more colonies, the more bacteria there were on the skin.

You will need

For each pair or group: six or more sterile nutrient agar plates; two large beakers; soap or soap solution; antiseptic solution such as Dettol, Solyptol or Cetavlon; marking pen; adhesive tape; an incubator (if available)

Planning your investigation

1. What will be your independent variable? You may wish to test different types of soap, different methods of hand washing, different lengths of time of hand washing, antiseptic solution vs antiseptic wipes, natural soaps vs synthetic soaps, or another factor.
2. What will be your dependent variable?
3. What variables will you need to control? How will you do this?
4. Draw up a suitable table in which to record your results.

Risk assessment

Bacteria can be dangerous and cause disease. Therefore, the lids on the plates should never be removed after they have been exposed. Plates should be autoclaved and disposed of following the investigation.

What to do

Follow the steps below for each of your tests.

1. Do not open the lid of the sterile culture plate until you are ready to press your fingers on to the surface. It is most important that exposure of the plates to the atmosphere be kept to an absolute minimum.
2. Press gently on the surface; do not push your fingers into the agar.
3. Replace the lid on the culture plate as quickly as possible.
4. Label the plate.
5. Tape the lid on to the plate with two pieces of adhesive tape so that it cannot be accidentally removed.
6. **Never remove the lid after the plate has been exposed.**
7. Incubate the plates upside down so that any moisture condensing on the lid of the plate cannot drip on to the nutrient medium.
8. At the end of the incubation period, count the number of bacterial colonies that have grown on the agar or calculate the area of the plate covered. You could also count the number of different species of bacteria. (Each one will have a different colour or texture.)
9. Autoclave the plates at 120°C for 20 minutes under 100 kPa pressure to make sure that any micro-organisms are destroyed.
10. Dispose of the plates with the lid still in place.

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Studying your results

Discuss the results of your investigation. Your discussion should include answers to the following questions.

1. Was your hypothesis supported or disproved?
2. What were some sources of error in your investigation? Did these affect its accuracy, reliability or validity?
3. How confident are you of your results? Why is this so?
4. What further investigations need to be made?
5. What improvement could be made to your experimental procedure?

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ACTIVITY 7.2 Investigating infectious disease transmission

An infectious disease is a pathogen that is passed from one host to another. These diseases can spread in several ways, including direct contact with an infected individual, indirect contact via surfaces or objects touched by an infected individual, and airborne droplets that result from infected individuals sneezing, coughing or laughing. The transmission of disease through these droplets depends on how close the infected individual and potential host are, as the droplets disperse and settle quickly. The common cold and influenza are typically transmitted through droplets in the air. Local health departments, the World Health Organization (WHO) and the Centres for Disease Control and Prevention (CDC) are responsible for monitoring infectious disease outbreaks. These agencies are responsible for identifying the source of outbreaks by tracking the routes of transmission. Over the past 100 years, these organisations, along with vaccine development and sanitation improvement, have effectively fought the spread of disease. Many of the infectious diseases that have historically been responsible for devastating epidemics have now been reduced or even eradicated.

Aim

To simulate a real-case scenario infectious disease transmission and identify patient zero
Time requirement: 45 minutes

You will need

1 screw-cap vial with solution (containing either 7 mL 0.001M hydrochloric acid or 7 mL 1M sodium hydroxide); phenol red indicator vial 15 mL; 1 plastic pipette; 4 96-well plates; marker; 1 index card; disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Sodium hydroxide can cause severe skin burns and eye damage.	Ensure that appropriate PPE is worn at all times.
Disposable gloves may pose allergy risk.	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.

What to do

1. Collect an index card, plastic pipette and a screw-cap vial containing solution. The solution in the vial represents bodily fluid. Your vial will be labelled with a number.
2. There are four well plates labelled 0, 1, 2 and 3. Locate your individual wells on the class well plates. These will be labelled with the number corresponding with the number on your vial.
3. Using a plastic pipette, remove some of the fluid from your vial and transfer five drops into your well on Well Plate 0.

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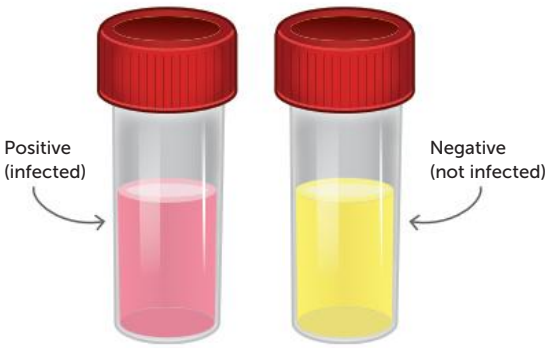
4 Select a partner, and record their name and vial number on your index card.
5 Using your plastic pipette, transfer five drops from your vial to your partner's vial. Return any liquid remaining in your pipette to your vial. Replace the vial cap and mix the solution by inverting it several times.
6 Using a plastic pipette, transfer five drops of liquid from your vial into your well on Well Plate 1.
7 Repeat steps 4 and 5 for the second and third exchanges, depositing your liquid into wells on Plates 2 and 3, respectively. Select a different partner for each round and complete each step before proceeding to the next exchange.
8 After all exchanges have been made, your teacher will add one drop of phenol red, an indicator solution that will determine if your vial has become 'infected'. Vials that turn red or pink are positive for the pathogen (infection), while vials that turn yellow are negative, indicating that your vial did not become infected.



9 Report if your vial tested positive. If so, share the names of the partners with whom you exchanged fluids.
10 Based on your individual results and the data from your classmates, try to identify which vial the infection spread from. Your teacher will add a drop of phenol red to each of the wells in the well plates. By observing which samples indicate a positive result in each round of transfers, you may be able to trace the spread of infection to the original source.
11 Copy and complete the table below to help you identify the source of infection. Once you have listed the positive vials and who they exchanged with, circle the numbers of the partners whose vials tested positive.

POSITIVE VIAL NUMBERS	1ST EXCHANGE PARTNER NUMBER	2ND EXCHANGE PARTNER NUMBER	3RD EXCHANGE PARTNER NUMBER

Studying your results

1 Who was patient zero?
2 After the three rounds of exchanges, how many vials tested positive? Calculate what percentage of your class this represents.
3 Graph how many students were infected after each round.

Discussion

1. If the class were divided into three groups of 10 at the start of this procedure and allowed to exchange only within their group, what would the transmission of the disease look like?
2. Did you know which vials were infected during the procedure?
3. Do you believe that an individual who does not show any signs of a disease can transmit it to others?
4. What is the importance of identifying patient zero in epidemics?
5. How does this simulation differ from the spread of disease in the real world, such as the spread of COVID-19? Explain.
6. List the appropriate measures that individuals should take to limit the spread of diseases.

Taking it further

Research past infectious disease epidemics. Your research should include:

- origins of the disease
- how the disease is transmitted
- typical incubation period
- symptoms/signs of the disease
- impact (i.e. death toll, cultural shifts, historical context)
- possible vaccines and treatments
- preventative measures.

ACTIVITY 7.3 Plotting a fever

Fever is when a person's body temperature is higher than the normal 37°C. It can result from injury, infection, toxins, reaction to a drug, or a number of other causes. At one time, it was thought that fever was harmful to the body and that everything possible should be done to reduce a high temperature. It is now known that, provided a person's temperature is not too high (over 40°C), fever can actually speed recovery.

The table shows the temperature recorded for a person who suffered a viral infection and recovered after about 10 days.

DAY	TIME	BODY TEMPERATURE (°C)
1	8 a.m.	37.1
	8 p.m.	37.4
2	8 a.m.	37.2
	8 p.m.	38.1
3	8 a.m.	38.6
	8 p.m.	39.2
4	8 a.m.	39.1
	8 p.m.	38.9
5	8 a.m.	39.2
	8 p.m.	39.3
6	8 a.m.	38.8
	8 p.m.	39.0
7	8 a.m.	39.1
	8 p.m.	38.7
8	8 a.m.	38.3
	8 p.m.	38.1
9	8 a.m.	37.7
	8 p.m.	37.4
10	8 a.m.	37.2
	8 p.m.	36.9
11	8 a.m.	37.1
	8 p.m.	37.2

What to do

1. Plot the data on a graph. Refer to Chapter 1 to review how to draw a graph correctly.
2. Describe what happened to the patient's temperature over the 11-day period covered by the data.
3. Calculate the patient's average temperature from 8 a.m. on day 3 to 8 p.m. on day 8.
4. During a fever, the body's 'thermostat' is set to a higher level. Explain how your graph illustrates this characteristic of a fever.

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ACTIVITY 7.4 Investigating the testing of animals in the manufacture of vaccines

Hold a class discussion on the scientific and ethical issues arising from the use of animals in the research and manufacture of vaccines. Assign the roles of interested parties to some of the members of the class, who will then assume that role in the discussion. The roles could include:

- a person suffering from a disease for which researchers are trying to develop a vaccine
- a spokesperson for an animal rights group
- a doctor specialising in immunology
- a member of the public opposed to the activities of animal rights groups
- an employee of the health department responsible for control of infectious diseases
- a scientist researching new vaccines
- a person opposed to the use of vaccines because of the risks involved.

As well as moral and ethical issues, the discussion could consider questions such as these.

1. Why do people's opinions differ about what activities should be allowed in the development and manufacture of vaccines?
2. How can society best consider the wide range of views that people hold on these issues?
3. Who should be allowed to decide whether testing on animals should be permitted?
4. What are the responsibilities of the scientists who use animals for their testing programs?
5. Who should set standards for laboratories and researchers that use animals for the manufacture and testing of vaccines?

After listening to the opinions expressed during the discussions, prepare a list of arguments for and against the use of animals for the manufacture and testing of vaccines.

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ACTIVITY 7.5 Investigating antibiotic resistance

Antibiotics are molecules that are produced by bacteria and fungi as a defence against other microbes. Penicillin was a revolutionary discovery for the human race in the 20th century. Along with other antibiotic discoveries, penicillin suddenly became a weapon against an invisible enemy. Antibiotics have been harnessed by scientists and medical professionals to treat diseases and save lives. Since that first discovery of antibiotics, they have been developed for use against the broad range of pathogenic microbes, each with their strengths and weaknesses. Unfortunately, this weapon has become dulled in the past decade as overuse has led to antibiotic resistance. Antibiotic resistance results from certain bacteria evolving to become resistant to the antibiotics that have been used to fight them. As a result, antibiotic medicines are not able to fight certain bacteria as effectively, and medical professionals have been forced to find alternative solutions. Not all antibiotics work against all bacteria, and knowing which bacteria are susceptible is essential to finding the best treatment for disease.

Aim

To investigate antibiotic effectiveness against common bacteria strains
Time requirement: 45 minutes

You will need

Escherichia coli broth culture; *Staphylococcus epidermidis* broth culture; 4 nutrient agar plates; 2 sterile pipettes; 2 disposable spreaders; 2 Mastring antibiotic discs; measuring ruler or callipers; sticky tape; marker; ethanol or bleach; sterile forceps; incubator; Bunsen burner; contaminated waste bag; disposable gloves

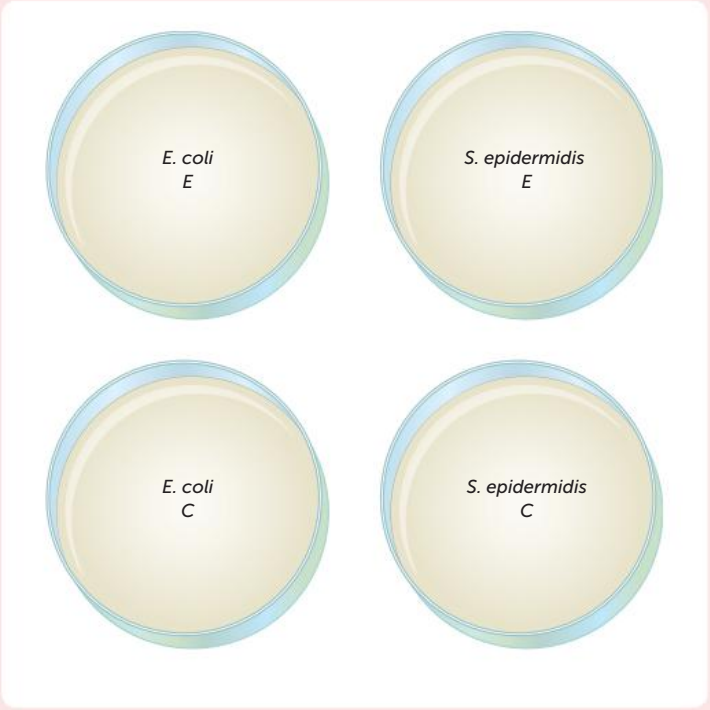
Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
While lab strains are usually harmless, bacteria may cause disease, so assume them to be pathogenic.	Wear lab coats, safety glasses and gloves; wash hands thoroughly at end. Decontaminate benches before and after activity. Flood spills with bleach.
Micro-organisms will grow on the agar plates.	Do not open plates once they are securely taped. Dispose of plates appropriately after autoclaving.
Disposable gloves may pose allergy risk.	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.

What to do

Note: To use aseptic technique, wipe your bench down with ethanol (or bleach) and keep your work near the Bunsen burner to take advantage of the updraught the flame will create to waft potential contaminants away from your materials.

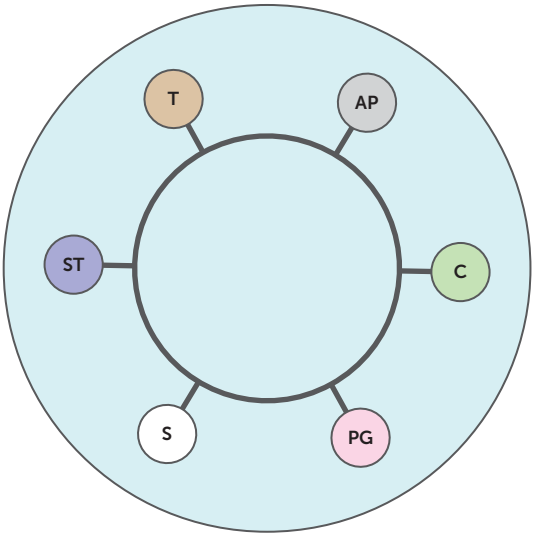
1 Label the bottom of your four agar plates with your name and the date. Label two plates *E. coli* and two plates *S. epidermidis*. Label one plate of each type of bacteria with 'E' for experiment and label the other 'C' for control.



2 Using a sterile plastic pipette, transfer one drop of the *E. coli* bacterial broth on to the surface of the agar on your two *E. coli* plates.
3 Working close to the Bunsen burner, use a spreader to spread the bacterial broth over the plates evenly. If you are using a glass spreader, pass it through the flame of the Bunsen burner before each use.
4 Replace the lids on the plates immediately to avoid contamination.
5 Repeat steps 2 to 4 for *S. epidermidis*, using a new sterile plastic pipette and spreader.

6 The next step is to apply the Mastring to each of the experiment plates. Wait 10–15 minutes before applying the Mastring to ensure that bacteria have a chance to grow.

7 To apply the Mastring, flame your forceps and allow them to cool before picking it up. Place it in the middle of your plate and push (very gently) with the forceps to help it stay in place. Each lobe of the Mastring is impregnated with a different antibiotic; use the code below or the one on the packet to differentiate them.



The symbols indicate antibiotics as follows:

- **AP** – AMPICILLIN (grey)
- **S** – STREPTOMYCIN (white)
- **C** – CHLORAMPHENICOL (green)
- **ST** – SULPHATRIAD (mauve)
- **PG** – PENICILLIN G (pink)
- **T** – TETRACYLINE (brown)

8 Repeat steps 6 and 7 for the other experiment plate, flaming the forceps between each application.

9 Seal all four plates with sticky tape and incubate for 24 hours at 37°C, upside down so that the agar is at the top.

10 Wipe your bench down with ethanol and clean your hands thoroughly.

11 Dispose of all materials safely in a contaminated-waste bag.

12 The next day, observe for the presence or absence of growth near the disc and measure the diameter of any zones of inhibition. Record your results and contribute to the class data pool.

Studying your results

1 Draw a diagram of what you see on each plate. Include labels.

2 Copy and complete the table below with the results of your experiment.

ANTIBIOTIC	DIAMETER OF ZONE OF INHIBITION (MM)	
	ESCHERICHIA COLI	*STAPHYLOCOCCUS EPIDERMIDIS*
Ampicillin		
Streptomycin		
Chloramphenicol		
Sulphatriad		
Penicillin		
Tetracycline		

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3 Calculate the class average diameter of the zone of inhibition for each antibiotic and copy and complete the table below with the results of your experiment.

ANTIBIOTIC	AVERAGE DIAMETER OF ZONE OF INHIBITION (MM)	
	ESCHERICHIA COLI	*STAPHYLOCOCCUS EPIDERMIDIS*
Ampicillin		
Streptomycin		
Chloramphenicol		
Sulphatriad		
Penicillin		
Tetracycline		

Discussion

1 Explain the function of the control plate in the experiment. How could a control plate be helpful in the event there is no growth on the experiment plate?
2 What were four variables that you kept constant in this experiment? How did you control them?
3 Why is it important to pool data from the class results and find the average zone of inhibition for each antibiotic?
4 What is a zone of inhibition? How were they created in your experiment?
5 Which antibiotic had the greatest zone of inhibition? Explain why this might be.
6 Did your individual results differ from the class results? If so, suggest possible reasons.
7 Which antibiotic would be most suitable to treat an infection by *Staphylococcus epidermidis*?
8 Which antibiotic would you use if you were unsure of the pathogen in an infection? Explain your answer.
9 Did your results show any signs of antibiotic resistance?
10 Discuss the effects that antibiotic resistance has on medical treatment.
11 Why have antibiotics become a less effective treatment for infection in recent years?

Taking it further

Test the efficacy of natural antibiotics on similar bacteria.

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CHAPTER 7 SUMMARY

- A pathogen is a disease-causing microorganism such as a bacterium or virus.
- A communicable, or infectious, disease can be transmitted from person to person.
- Bacteria are single-celled organisms that usually have a cell wall but not an organised nucleus or membrane-bound organelles. Most bacteria are harmless, or even beneficial. However, some are pathogens. Bacteria are classified by their shape and can be identified after growing on an agar plate of nutrient medium and then being viewed under a microscope.
- Viruses contain DNA or RNA within a protein coat. Some viruses also have an external envelope.
- Viruses cannot reproduce themselves. Instead, they infect a cell and use the cell to produce many copies of the virus, which are then released to infect more cells.
- Transmission of pathogens can occur by direct contact or ingestion, via body fluid, droplets or airborne particles, or through another animal called a vector.
- Non-specific defences, including external defences, protective reflexes, phagocytosis, inflammation, fever and the lymphatic system, are for all pathogens; while specific defences, including cell-mediated responses and antibody-mediated responses, are for one specific pathogen only.
- External defences stop a foreign particle entering the body. They include the skin, mucus, hairs and cilia, acid, lysozyme, cerumen and the movement of fluid.
- Protective reflexes are automatic responses to protect the body by eliminating the foreign particle or pathogen. They include sneezing, coughing, vomiting and diarrhoea.
- Phagocytosis involves a phagocyte engulfing and digesting the pathogen. Macrophages, neutrophils and dendritic cells are phagocytes.
- Inflammation occurs when tissue is damaged and causes heat, redness, swelling and pain. Mast cells release histamine, which increases blood flow to the damaged area and causes fluid to leak out of the vessels into the surrounding tissue. Heparin is also released and prevents the blood clotting. Phagocytes are attracted to the area and consume the pathogen, removing the cause of infection, allowing new cells to form and the tissue to heal.
- Pyrogens, such as Interleukin-1, can cause the set point for body temperature to increase. This means that homeostatic mechanisms will result in an increased body temperature. While this may help by inhibiting the growth of the pathogen, it is dangerous if it gets too high.
- The lymphatic system plays a role in non-specific responses, with lymph nodes filtering lymph and macrophages destroying pathogens.
- B-cells and T-cells are lymphocytes responsible for the body's specific defences by responding to antigens (large molecules that are capable of triggering an immune response).
- Antigen-presenting cells, such as dendritic cells and macrophages, engulf and digest the pathogen, then display the antigen on their surface to present to lymphocytes.
- The humoral response, or cell-mediated immunity, occurs when B-cells for a specific antigen are activated, forming a clone. Some of the clones become plasma cells, which produce antibodies; the rest become memory cells.
- Antibodies are specific to a particular antigen. They combine to form an antigen–antibody complex which can respond to pathogens by inactivating them, preventing them from entering cells, causing agglutination or increasing the chance of phagocytosis.

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- Memory cells remain in the body and are quick to respond to future exposures to the antigen.
- The first exposure to an antigen is the primary response. There is a delay before the level of antibodies is sufficient. Upon a second exposure, memory cells allow the body to respond much more quickly and produce greater levels of antibodies that last longer. This allows the body to remove the pathogen before it causes the disease.
- Cell-mediated immunity occurs when T-cells are activated by a B-cell or macrophage presenting the antigen to the T-cell. The T-cell enlarges, multiplies and produces a clone, which may become a killer T-cell or a helper T-cell. Killer T-cells inactivate the pathogen by releasing substances that destroy it. Helper T-cells act indirectly by causing an increase in phagocytosis. If the response is too great, suppressor T-cells are produced, which inhibit T-cell and B-cell activity. Memory cells are also produced, which remain in the body and respond quickly if the same pathogen enters the body again.
- Immunity may be natural or artificial.
- If immunity is due to receiving antibodies, without the body responding to an antigen, it is passive immunity – for example, a baby receiving antibodies in breast milk. If immunity is due to the body responding to antigens, it is active immunity – for example, being exposed to a pathogen.
- Vaccinations can lead to immunisation by artificially introducing pathogens and allowing the body to develop immunity. Vaccines do not cause the disease as the antigen is introduced without the active pathogen. For example, the pathogen may be inactivated (attenuated) or dead, or the vaccine may contain an inactivated toxin or only part of the pathogen.
- Recombinant DNA is being investigated to make vaccines by adding the DNA responsible for the antigen to harmless bacteria. This should allow safer, more effective vaccines to be developed.
- Booster shots of vaccines are often needed to utilise the secondary response to ensure that the levels of antibodies and memory cells are sufficient to protect the body from disease.
- If enough people in a population are immune to a disease, they can protect the rest of the population by making the spread of the disease less likely. This is herd immunity.
- There are many factors to consider in relation to vaccinations. These include allergic reactions, ethical concerns and religious beliefs.
- Antibiotics fight infections, particularly those due to bacteria, by destroying the cell wall or membrane, or stopping their reproduction.
- Some bacteria have evolved to become resistant to antibiotics. This evolution has been hastened by incorrect use of antibiotics.
- Antiviral drugs treat viral infections. They are harder to develop, as viruses invade the host cells. Therefore, killing a virus also affects its host cells.

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CHAPTER 7 GLOSSARY

Active immunity Immunity produced by the body manufacturing antibodies against a foreign antigen

Agglutination The clumping together of micro-organisms or cells

Antibiotic A chemical able to inhibit the growth of, or to kill, micro-organisms, particularly bacteria

Antibody A substance produced in response to a specific antigen; combines with the antigen to neutralise or destroy it

Antibody-mediated immunity *see* humoral response

Antigen Any substance capable of causing the formation of antibodies when introduced into the tissues

Antigen–antibody complex A compound formed when an antibody combines with an antigen

Antigen-presenting cells Phagocytic cells that digest pathogens and present the antigen to lymphocytes; include dendritic cells and macrophages

Antiviral drug A drug used for the treatment of viral infections

Artificial immunity Immunity produced by giving a person an antigen, which triggers the immune response, or by giving them antibodies to an infecting antigen

Attenuated Describes micro-organisms that have been reduced in virulence

Bacteria Unicellular, prokaryotic organisms with a cell wall but lacking membrane-bound organelles and an organised nucleus; singular: bacterium

Bactericidal antibiotic A drug used to treat bacterial infections by killing the bacteria

Bacteriophage A virus that infects bacteria

Bacteriostatic antibiotic A drug used to treat bacterial infections; it does not kill the bacteria but stops them reproducing

B-cell A type of lymphocyte that develops into either a plasma cell that produces antibodies or a memory cell

Broad-spectrum antibiotic An antibiotic that affects many types of bacteria

Cell-mediated response The part of the immune response in which T-cells attach to antigens to destroy them; also called cellular immunity

Cerumen Ear wax; secreted by special glands near the opening of the ear canal

Cilia Hair-like projections from a cell; they beat rhythmically to move material across a tissue surface; singular: cilium

Clone A group of cells with the same genetic characteristics

Communicable disease A disease passed from one person to another by infection with micro-organisms; also called an infectious or transmissible disease

Complement system A system of proteins produced by the liver that enhance the activity of antibodies and phagocytes

Contagious A disease passed on by direct human contact

Cytokines Small proteins that are released in response to antigens and act as messengers in the immune response

Cytotoxic T-cells *see* killer T-cell

Dendritic cell An antigen-presenting cell, named due to the branch-like extensions from the cytoplasm

Fever An elevation of body temperature above the normal level of 37°C

Helper T-cell A type of T-cell that, among other things, enhances antibody production by B-cells

Heparin A substance that helps to prevent blood clotting

Herd immunity A type of 'group' immunity that occurs when such a high proportion of people in a population are immunised that those who are not immune are protected

Histamine A substance released in response to injury to cells; it results in an increase in blood flow

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Humoral response A response triggered by foreign substances or micro-organisms entering the body, involving B-cells and the production of antibodies; also known as antibody-mediated immunity

Immune response A response triggered by foreign substances or micro-organisms entering the body

Immune system Different types of cells that occur in most organs of the body and that protect against foreign organisms, alien chemicals and abnormal cells

Immunisation Programming the immune system so that the body can respond rapidly to infecting micro-organisms

Immunity Resistance to infection from invading micro-organisms

Immunoglobulin A particular group of proteins; antibodies are immunoglobulins

Infectious disease *see* communicable disease

Inflammation The response to damage to a tissue; involves swelling, heat, pain and redness in the affected area

Interferon Any of several proteins that are produced by cells as a defensive response to viral infection, preventing the replication of the virus

Interleukin-1 A pyrogen produced primarily by macrophages

Killer T-cell A type of T-lymphocyte able to kill cells that are damaged or infected with viruses or bacteria; also called cytotoxic T-cell

Leucocyte A white blood cell; also spelt leukocyte

Lymphatic system A system of vessels that drain excess fluid from the tissues; also called the lymph system

Lymphocyte A white blood cell that is responsible for the immune response

Lymphoid tissue Tissue containing many lymphocytes and macrophages; found mostly in the lymph nodes but also in the bone marrow, tonsils, spleen and thymus

Lysozyme An enzyme that kills bacteria; found in tears, saliva and perspiration

Macrophage A phagocytic cell derived from a monocyte (a type of white blood cell)

Mast cell A type of cell found in loose connective tissue; involved in the inflammatory response

Memory cell A type of cell that recognises an antigen to which the body has previously been exposed

Monocyte A type of leucocyte found in the blood that migrates into damaged tissue and forms macrophages

Mucous membrane An epithelial tissue that secretes mucus and lines many body cavities

Mucus A slippery, stringy substance produced by mucous membranes

Multiple drug resistance Resistance of some strains of bacteria to most of the available antibiotics

Narrow-spectrum antibiotic An antibiotic that affects only a particular type of bacteria

Nasal cavity The large air-filled cavity above and behind the nose

Natural immunity Immunity that occurs without any human intervention

Neutrophil A granulated leucocyte with a multilobar nucleus that is phagocytotic

Non-self antigen Any compound foreign to the body that triggers an immune response

Non-specific defence Defence of the body that acts against all pathogens

Passive immunity Immunity produced by the introduction of antibodies from another person

Pathogen A disease-causing organism; often referred to as a pathogenic organism

Phagocyte Cells that are able to engulf micro-organisms and cell debris

Plasma cell A cell that develops from a B-cell and produces antibodies

Plasmid In a bacterial cell, small circular strands of DNA distinct from the main bacterial genome; composed of only a few genes and able to replicate independently within cells

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Primary response The response of the immune system to the first exposure to an antigen

Prokaryote A single-celled organism lacking a distinct nucleus or specialised organelles

Pyrogen A substance that results in a fever

Recombinant DNA Synthetic DNA; made by inserting genes from one source into a DNA molecule from a different source

Sebum An oily, waxy secretion from the sebaceous glands

Secondary response The response to a second or subsequent exposure to an antigen; the secondary response is faster and more intense than the primary response

Self-antigen Any large molecule produced in a person's own body; does not cause an immune response in that person

Specific defence Defence of the body that is directed against a specific pathogen

Suppressor T-cell A type of T-cell that helps to slow down the immune response

Sweat The liquid produced by the sweat glands on the skin

T-cell A lymphocyte that can differentiate into a number of different kinds of cell, all of which are involved in cell-mediated immunity

Total drug resistance The resistance of some strains of bacteria to all antibiotics

Toxoid A toxin from a pathogenic organism that is altered so that it is no longer toxic

Vaccination The introduction of antigens to a person so that they acquire immunity without suffering from the illness

Vaccine An antigen preparation used in artificial immunisation

Vasodilation An increase in the diameter of arterioles, increasing the flow of blood through them

Vector An agent such as an insect capable of transferring a disease-causing organism from one person to another

Virulence The disease-producing power of a micro-organism

Virus An infectious agent, too small to be seen with a light microscope, consisting of a protein sheath surrounding a core of nucleic acid; viruses are totally dependent on living cells for reproduction

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CHAPTER 7 REVIEW QUESTIONS

Recall

1 Define 'communicable disease' and name five examples.

2 List the external defences that prevent the entry of pathogenic organisms into the body.

3 a How do protective reflexes help to defend the body from infection by pathogenic organisms?
  b List four reflexes that help to protect against infection.

4 In the inflammatory response, describe the role of:
  a mast cells
  b histamine
  c heparin
  d phagocytes.

5 How is fever during the course of an infection thought to be beneficial?

6 Why is the immune response said to be a specific response?

7 a What is an antigen?
  b Explain the difference between self-antigens and non-self antigens.

8 List the ways in which the antigen–antibody complex helps to overcome the effects of invading micro-organisms.

9 List the ways in which killer T-cells and helper T-cells can deal with an invading antigen.

10 a How can passive immunity be gained artificially?
  b How can active immunity be acquired naturally?

11 a What is a vaccine?
  b Describe three ways in which older types of vaccines are produced.
  c What new methods are being trialled to produce vaccines?
  d List the risks associated with the use of vaccines.

12 Explain the difference between:
  a an antibiotic and an antiviral
  b a bactericidal and a bacteriostatic antibiotic
  c a broad-spectrum and a narrow-spectrum antibiotic.

Explain

13 Explain the difference between:
  a a pathogen and a vector
  b RNA viruses and DNA viruses
  c bacteria and bacteriophages.

14 Explain the importance of phagocytes in defence against disease.

15 Explain what causes the four signs of inflammation.

16 Explain the difference between:
  a natural and artificial immunity
  b active and passive immunity.

17 Why is the secondary immune response so much faster than the primary response?

18 Why is it rare to get a disease such as measles or chickenpox more than once?

19 Explain how T-cells are able to produce immunity.

Apply

20 a Bacteria were first detected in 1683, but viruses were not detected until 1938. Suggest why this happened.
  b List four differences between bacteria and viruses.

21 Explain how coughing into your elbow can help reduce the spread of disease.

22 Draw a flow chart showing the events that occur in an inflammatory response.

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23 Explain why someone with an infected toe may develop a lump in the groin.

24 During a fever, people often have severe chills and can shiver uncontrollably even though their temperature is above normal. Explain how this is thought to come about.

25 Compare and contrast antigens and antibodies.

26 Draw a flow chart to show how cell-mediated immunity is activated.

27 Typhoid is caused by a bacillus. To make a positive diagnosis of typhoid, a sample of the patient's blood is taken and mixed with typhoid bacilli. If the bacilli agglutinate, the patient has typhoid.

- **a** Why is this a positive diagnosis for the disease?
- **b** Could the person be suffering from some other disease?

28 A person was prescribed an antibiotic for a bacterial infection of the throat. While taking the antibiotic tablets, the patient developed a bacterial infection in their big toe. Explain why the antibiotics that the patient was taking for the sore throat did not prevent the growth of bacteria in the toe.

Extend

29 During the COVID-19 pandemic in 2020, there was debate about the effectiveness of the general public wearing masks. Discuss both sides of this debate.

30 The Russian composer Tchaikovsky died of cholera during an epidemic in Moscow in 1893. It is believed that Tchaikovsky drank unboiled water during the epidemic, some think in a deliberate attempt to commit suicide. Why would drinking unboiled water increase the risk of cholera infection?

31 The body's immune system does not normally react against its own antigens – the body is said to have tolerance for its own antigens. However, sometimes this tolerance breaks down. Conduct research to find out:

- **a** what autoimmune diseases are
- **b** what causes these diseases
- **c** how autoimmune diseases are treated.

32 Investigate and report on the issues surrounding the use of vaccines to protect against human papilloma virus (HPV). Ensure that you provide a balanced discussion of both sides of the subject.

33 Reye's syndrome (pronounced 'rise') is a serious disorder that sometimes occurs in children after a viral infection such as chickenpox or the flu. It was first recognised as a distinct disorder in 1963 by R. Douglas Reye, an Australian pathologist. Reye's syndrome mainly affects children between the ages of 4 and 16 years, and statistics show that it can be triggered by the use of drugs that reduce fever, such as aspirin. Use the Internet to research Reye's syndrome, including its:

- **a** causes
- **b** signs and symptoms
- **c** long-term consequences
- **d** frequency
- **e** prevention.

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8 TECHNOLOGY IS USED TO TREAT DISEASES

UNIT 3 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including the collection of data related to homeostasis and the use of models of disease transmission, safely, competently and methodically for the collection of valid and reliable data
- represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions

SCIENCE AS A HUMAN ENDEAVOUR

- synthetic hormones may be developed to control or treat endocrine dysfunction, including diabetes mellitus, hypothyroidism and hyperthyroidism, to improve the quality of life for individuals
- gene therapy can be used to treat a range of diseases, including diabetes mellitus
- hormones and vaccines are developed using recombinant DNA and associated biotechnological techniques
- cell replacement therapy has the potential to treat nervous system disorders including Alzheimer's and Parkinson's diseases

Source: School Curriculum and Standards Authority, Government of Western Australia

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Do you eat bread, cheese or yoghurt? If so, you are consuming foods that are produced by biotechnology. **Biotechnology** uses cellular processes to make products that are of use to humans. For thousands of years people have been using yeasts to make bread and alcohol, and bacteria to make cheese and yoghurt.

Shutterstock.com/Jeroen Nieuwenhuisen

FIGURE 8.1 Cheese making uses ancient biotechnology methods

The science of cheese

Modern biotechnology has dramatically expanded the range of techniques and products that can be used to improve human welfare. Improvements in the treatment and prevention of disease, food production, production of clean energy and enhanced efficiency of manufacturing processes are all resulting from advances in biotechnology.

Therefore, the definition of biotechnology has more recently been expanded to include genetic testing, gene manipulation, cell replacement therapies and tissue engineering. Some of the methods and outcomes of modern biotechnology are described in this chapter.

8.1 RECOMBINANT DNA

Many methods of modern biotechnology utilise our understanding of DNA and apply it to specific processes.

DNA

You learnt about DNA in *Human Perspectives ATAR Units 1 & 2*; however, we will review it briefly here. DNA (deoxyribonucleic acid) is found in the cells of all organisms, usually in the nucleus, but there is also some in the mitochondria and, in some organisms, in the cytosol. All DNA molecules consist of two strands of nucleotides. Each nucleotide is made up of a deoxyribose sugar molecule, a phosphate group and a nitrogenous base. When these nucleotides join together, it makes a backbone of alternating deoxyribose sugar and phosphate with nitrogen bases branching at each sugar molecule. The bases from two strands are attracted to one another by hydrogen bonds, and this forms cross-links between the two strands. The DNA molecule is twisted into a spiral known as a double helix.

There are four different nitrogen bases in a DNA molecule: adenine (A), thymine (T), cytosine (C) and guanine (G). The base pairs are complementary; adenine will only pair with thymine, and cytosine will only pair with guanine. The order in which the nitrogen bases occur in the DNA molecule is the genetic information that determines the structure of the cell and the way it functions.

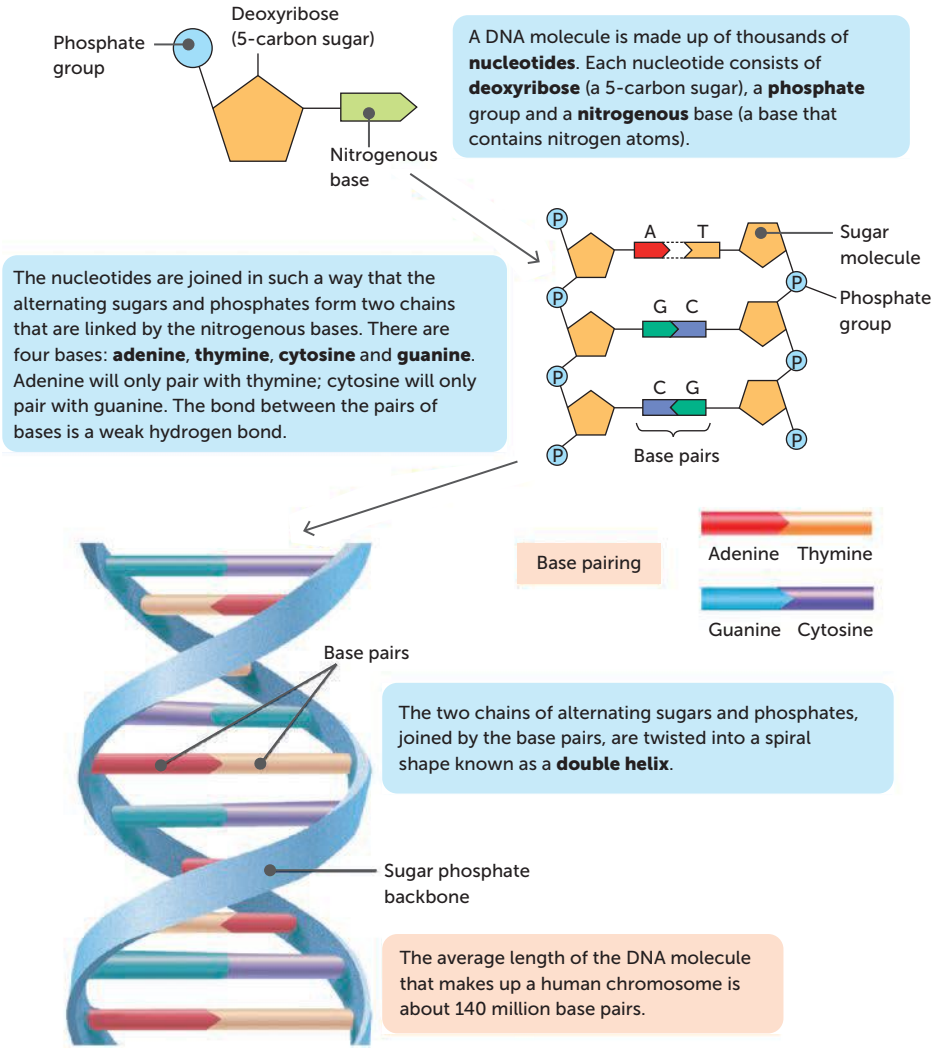

FIGURE 8.2 DNA consists of two strands of alternating sugars and phosphates with pairs of nitrogen bases forming cross-links between the chains. The two strands are twisted into a double helix

Recombinant DNA technology

Scientists have been modifying the DNA of organisms for a long time. By selecting which male and female organisms are crossed to produce offspring, we are increasing the chance of certain genes being present in the DNA of the next generation. If parents with desired traits are chosen, we can increase the chance of the gene for those phenotypes being passed on. And when parents without undesirable traits are chosen, there is a decreased chance of the genes for that phenotype occurring in the next generation. In this manner, we can either increase or decrease the incidence of certain genes. This process is known as **artificial selection** or **selective breeding**.

Artificial selection is quite a slow and inefficient process. Genes are passed on by chance and it is necessary to wait for the next generation to mature before knowing the outcome.

Shutterstock.com/Nature Art

FIGURE 8.3 Pugs are an example of selective breeding being used to increase the occurrence of genes for desirable traits

An alternative process is **genetic engineering,** which involves the artificial modification of DNA. This is also known as **recombinant DNA technology**. In this process, DNA is either added or removed from a cell. The DNA produced is called **recombinant DNA** and the organism is a **genetically modified organism (GMO)**. This technology has a wide range of possible uses, including introducing genes for desired traits into organisms, using harmless bacteria to produce proteins, and replacing faulty genes.

AAP Photos/Erik de Castro/Reuters

FIGURE 8.4 Golden Rice is named for its golden colour due to the presence of beta-carotene

Some applications of genetic engineering involve DNA from one species being introduced into a different species. The organism produced is a **transgenic organism**. The aim of transgenic organisms is to introduce a trait that is not normally present. All transgenic organisms are GMOs, but not all GMOs are transgenic. One example of a transgenic organism is Golden Rice, which was developed to address vitamin A deficiency in developing countries. The deficiency kills approximately 670 000 children under the age of five every year. Golden Rice was produced by introducing a gene from maize and a bacterium found in soil into rice. This allows the rice to produce beta-carotene, which the human body can use to synthesise vitamin A.

Golden Rice
This website provides more information about Golden Rice.

Key concept

Recombinant DNA technology, or genetic engineering, involves artificially changing DNA and produces a genetically modified organism. If this organism has DNA from another species, it is called a transgenic organism.

Development of recombinant DNA technology

Stanley Norman Cohen and Herbert Boyer invented the recombinant DNA technique in 1973. Their technique was to isolate and amplify genes or DNA segments and insert them into a bacterial cell, creating a transgenic bacterium. The introduced genes become part of the transgenic organism's DNA and can be passed on from one generation to the next.

Transgenic or GMO?
This website explains the difference between transgenic organisms and genetically modified organisms in more detail.

Restriction enzymes

For genetic engineering to be possible, the gene for the desired trait must be identified and then isolated. Next, the DNA receiving the gene must be 'opened', and the gene is then added to the recipient and joins its DNA.

A key breakthrough in genetic engineering involved viruses that infect bacterial cells. These are called **bacteriophages**, or **phages**. It was discovered that certain enzymes in bacteria are able to restrict the duplication of bacteriophages by cutting up the viral DNA. Scientists found that these enzymes always cut the DNA at a point where there is a certain sequence of bases. This sequence is known as a **recognition site** (or **recognition sequence**), and the enzyme that cuts the DNA is a **restriction enzyme** because it restricts the duplication of bacteriophages.

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Restriction enzymes are examples of **endonucleases**, enzymes that cut within a DNA molecule by separating two nucleotides. Some restriction enzymes produce a straight cut at the sequence, while others produce a staggered cut.

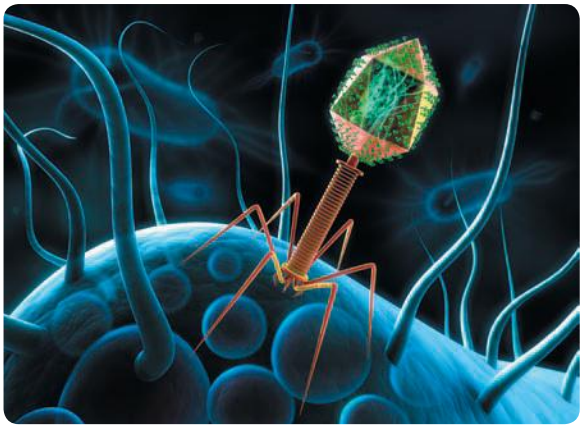
- A **straight cut** is when the restriction enzyme makes a clean break across the two strands of DNA to produce a blunt end. A **blunt end** is when both strands terminate in a base pair.
- A **staggered cut** results in fragments with sticky ends. A **sticky end** is a stretch of unpaired nucleotides in the DNA molecule that overhang at the break in the strands.

Recognition sites are four to eight base pairs in length and are **palindromic**, meaning that they have the same sequence when read both forward and backwards. This, along with the complementary nature of the bases, means that the same sequence occurs on both strands within the recognition site. Therefore, both strands will be cut, resulting in the DNA molecule forming two segments.

Each restriction enzyme will:

- recognise a certain base sequence
- cut at a certain point.

Both of these factors contribute to the type of cut. For example, the XmaI enzyme and SmaI both have the same recognition site, 5′-CCCGGG-3′. XmaI cuts between the first and second nucleotides and produces sticky ends, while SmaI cuts between the third and fourth nucleotides and produces blunt ends.



Shutterstock.com/peterschreiber.media

FIGURE 8.5 A digital image of a bacteriophage infecting a bacterium

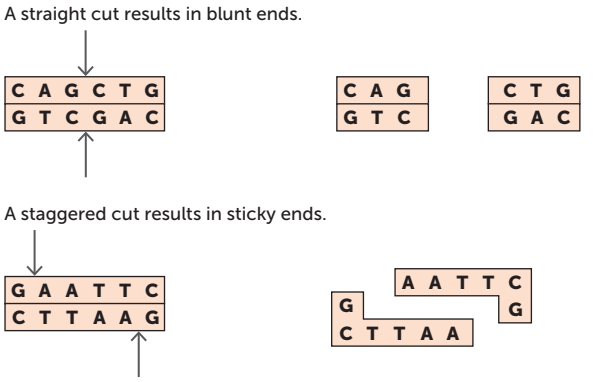
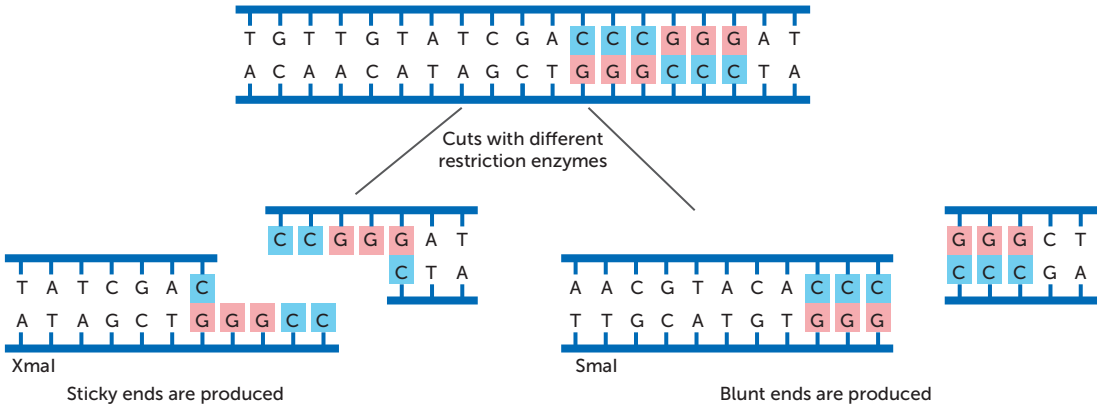


FIGURE 8.6 Cuts in DNA strands produced by restriction enzymes

FIGURE 8.7 The recognition site and position of the cut determine the types of ends produced by restriction enzymes



Sticky ends are so named because of their ability to combine with sections of DNA that have a complementary ending. These are very useful in recombinant DNA technology as they allow a single-stranded overhang from one DNA fragment to be paired with any other piece of DNA that has a corresponding sequence. This DNA could be from the same or a different organism.

Table 8.1 lists some restriction enzymes. The name of each enzyme reflects its origin.

- The first letter of the name comes from the genus of the bacterium from which it was isolated.
- The second two letters come from the species.
- The next letter refers to the strain of the bacterium.
- The roman numerals represent when the enzyme was isolated, where I is the first enzyme isolated, II is the second enzyme isolated etc.

For example, EcoRI is the first restriction enzyme isolated from the RY13 strain of the bacterium *Escherichia coli*, while HindIII is the third enzyme isolated from the R(d) strain of the *Haemophilus influenzae* bacterium.

Restriction enzymes
Use the simulation to view the cuts produced by different restriction enzymes.

Activity 8.1
Investigating restriction enzymes

TABLE 8.1 Examples of restriction enzymes

ENZYME	RECOGNITION SITE	BACTERIAL ORIGIN
BamHI	G↓G A T C C C C T A G↑G	*Bacillus amyloliquefaciens*
EcoRI	G↓A A T T C C T T A A↑G	*Escherichia coli*
HindIII	A↓A G C T T T T C G A↑A	*Haemophilus influenzae*
TaqI	T↓C G A A G C↑T	*Thermus aquaticus*
PvuII	C A G↓C T G G T C↑G A C	*Proteus vulgaris*

Key concept

Restriction enzymes cut DNA at a palindromic recognition site, producing either blunt ends or sticky ends in sections of DNA.

DNA ligase

Another major breakthrough in being able to modify genes was the discovery of an enzyme that was able to join, or recombine, separate pieces of DNA. This enzyme, found in the bacterium *Escherichia coli* (*E. coli*), was originally called a 'DNA-joining enzyme', but is now known as **DNA ligase**. Some version of DNA ligase is used by every living cell to 'glue' together short strands of DNA during replication, a process called **ligation**.

DNA ligase works by joining the phosphate group at the end of one strand to the sugar molecule at the end of another strand. For this to be possible, the complementary bases must first join by forming hydrogen bonds. Then the DNA ligase can join the backbone of each strand.

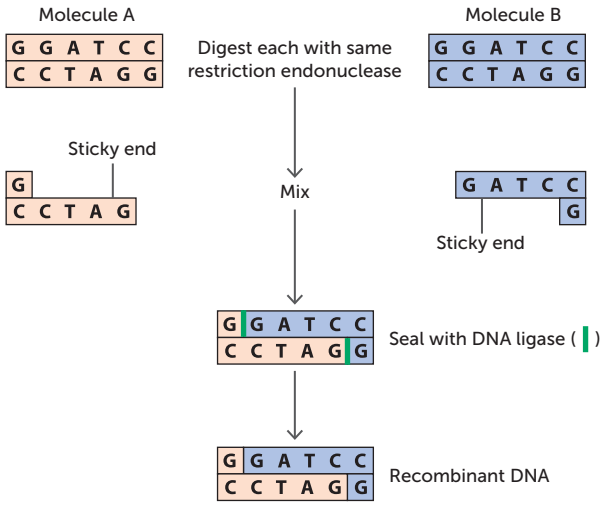


FIGURE 8.8 Making a recombinant DNA molecule

Activity 8.2
Investigating bacterial transformation

Key concept

DNA ligase is able to join sections of DNA.

Use of vectors

In genetic engineering, a **vector** is a DNA molecule that is used to carry DNA into a cell. The first step in producing an organism with recombinant DNA is to isolate the gene of interest. This gene is then inserted into a vector and cloned. This is achieved by:

1. identifying the desired gene
2. using a restriction enzyme to cut the DNA on either side of the gene
3. using the same restriction enzyme to cut the DNA of the vector
4. adding the desired gene to the vector
5. using DNA ligase to join the two sections of DNA.

Video on recombinant DNA
This website has a video showing how DNA recombination can be used in the manufacture of certain proteins.

In recombinant DNA technology, commonly used vectors are bacterial plasmids and bacteriophage viruses. **Plasmids** are usually circular, double-stranded units of cytoplasmic DNA, frequently found in bacteria, that are capable of replicating within a cell independently of the chromosomal DNA. The gene of interest is integrated into the plasmid or phage, and is referred to as recombinant DNA. Cloning of the vector then occurs so that numerous copies of the DNA are available to insert into the host cells. Once large quantities of the vector have been produced, they can be introduced into the selected host cells such as special bacterial, yeast or mammalian cells. These host cells will then produce the foreign protein using instructions in the gene in the recombinant DNA.

Bacteria into which the gene for insulin production has been introduced are now used in the manufacture of insulin for the treatment of diabetes.

Key concept

Vectors, such as plasmids or bacteriophages, can be used to transfer DNA into a host cell.

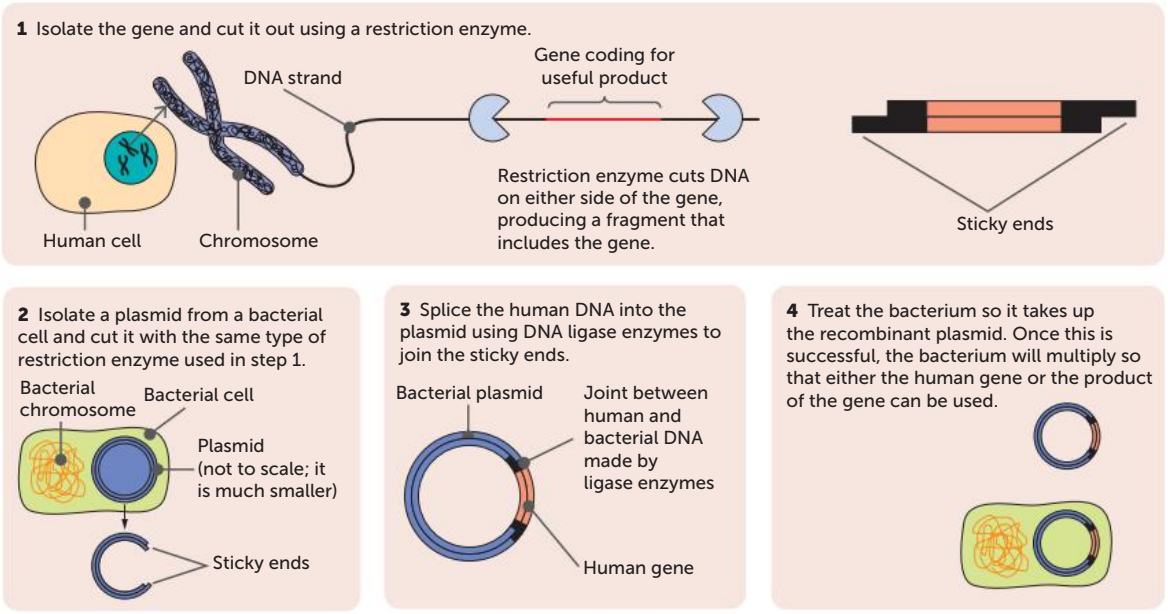


FIGURE 8.9 A simplified diagrammatic representation of recombinant DNA technology

TABLE 8.2 Terminology for recombinant DNA technology

TERM	DEFINITION
Blunt ends	The ends produced by a straight cut of a sequence of nucleotide bases
DNA ligase	An enzyme capable of combining two small components of single-stranded DNA into one single structure
Phage	Or bacteriophage; a virus that infects bacteria
Plasmid	A small circular strand of DNA distinct from the main bacterial genome; it is composed of only a few genes and is able to replicate independently within a cell
Restriction enzyme	An enzyme that cuts a strand of DNA at a specific sequence of nucleotides called the recognition site
Staggered cut	Produced when a restriction enzyme creates fragments of DNA with unpaired nucleotides that overhang at the break in the strands; called sticky ends
Straight cut	Produced when a restriction enzyme makes a clean break across the two strands of DNA so that the ends terminate in a base pair; called blunt ends
Sticky ends	The overhanging ends produced by a staggered cut of a sequence of nucleotide bases; can be called cohesive ends
Vector	A bacterial plasmid, viral phage or other such agent used to transfer genetic material from one cell to another

Examples of the use of recombinant DNA technology

Recombinant DNA technology has had an enormous impact on the diagnosis and treatment of diseases and genetic disorders. It has also enabled the manufacture of large quantities of pure protein for many medical products, including insulin, growth hormone, factor VIII and follicle-stimulating hormone (FSH). In the past these substances had to be extracted from people or animals, and they were often impure and/or of variable strength. One example was the transmission of the human variant of Creutzfeldt-Jakob disease (vCJD) by contaminated human growth hormone. This disease, a variant of 'mad cow disease', is a rare but fatal brain infection. There is evidence that some blood products used to produce the protein factor VIII were contaminated with vCJD.

Shutterstock.com/Varvara Vishnevska

FIGURE 8.10 GloFish®, the first commercially available transgenic organism, are a type of zebra fish that have been modified through the insertion of a gene that codes for the production of a protein that glows with a green fluorescence

Designer hens
This website shows how designer hens are able to lay eggs containing proteins to fight human diseases.

Recombinant DNA and vaccines

The first vaccine for human use that was produced using recombinant DNA technology was the hepatitis B vaccine, introduced in 1986. It was produced by inserting a gene from the hepatitis B virus into the cowpox virus.

Most of the vaccines currently being investigated are focused on using recombinant bacterium *E. coli* or cells from mammals, insects or yeast to produce protein antigens. These can be introduced to the body, where they will elicit an immune response. Vaccines produced using recombinant DNA are known as **recombinant vaccines**.

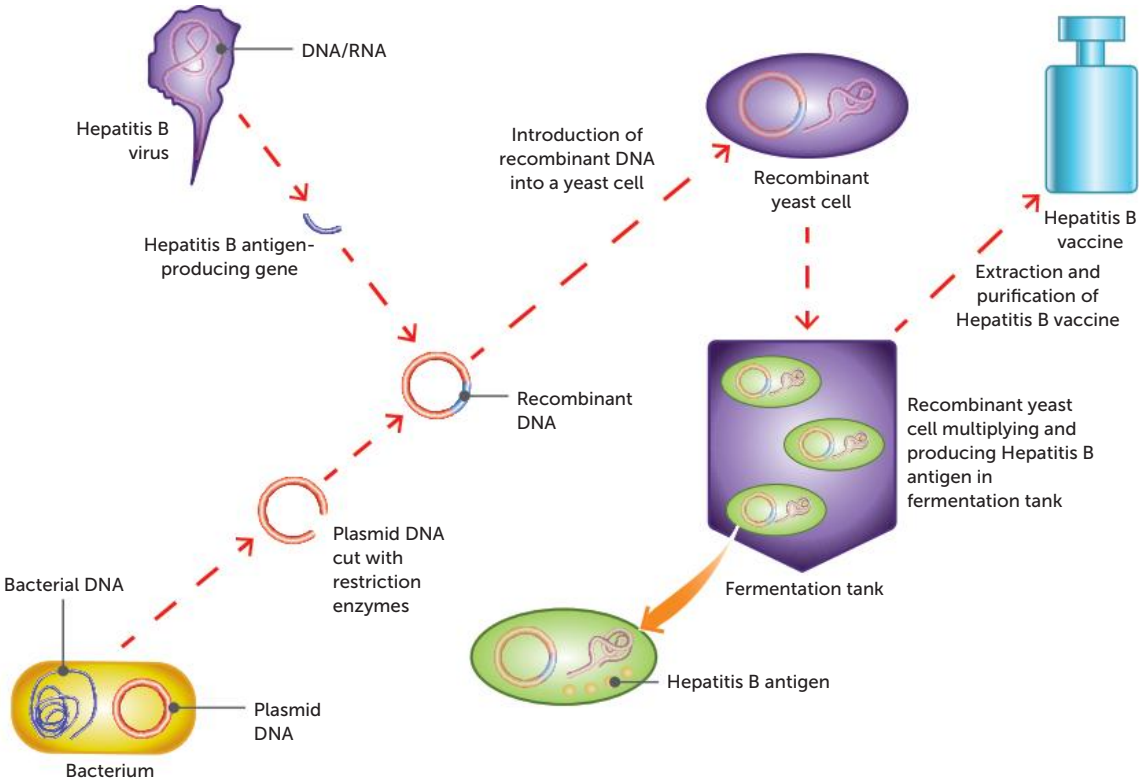


FIGURE 8.11
Recombinant yeast cells are used to produce a vaccine for hepatitis B

Currently, the vaccine for hepatitis B is produced using recombinant technology. The gene for a surface antigen on the virus is isolated and added to a plasmid. The plasmid is introduced into a yeast cell. When the yeast cell divides, the new cells also contain the plasmid with the gene for the antigen. This gene allows the yeast cells to produce the antigen protein, which can be collected and purified.

Another vaccine produced by recombinant technology is for the human papilloma virus (HPV). It is produced in a very similar process, using recombinant yeast or insect cells. These cells produce proteins that are found in the coat of the virus and are collected to be used in the vaccine.

Another area of research is **DNA vaccines**. With recombinant vaccines, the antigen is produced and then introduced in the vaccine. Yet, with DNA vaccines the DNA for the antigen is introduced in the vaccine instead of the antigen itself. The DNA is incorporated into the host's cells, which will produce the antigen. The thought is that the antigen will then be expressed by the host cells, in a similar way to what happens during a viral infection.

Development of recombinant DNA vaccines does have disadvantages. It is very expensive, as the genes for the desired antigens must be located, cloned and expressed efficiently in a new vector. In addition to this financial deterrent to innovation, those involved in vaccine research must also be conservative. Because vaccines are used on large numbers of healthy people, many of whom are children, the safety of the product is paramount. Therefore, if a conventional vaccine is known to be safe, there is little incentive to develop a new one using genetic engineering.

Key concept

Recombinant DNA technology can be used to synthesise hormones and vaccines.

Questions 8.1

RECALL KNOWLEDGE

1 Define:
- **a** recombinant DNA technology
- **b** genetically modified organism
- **c** transgenic organism
- **d** vector
- **e** palindromic
- **f** plasmid
- **g** bacteriophage.

2 Draw a labelled diagram to show the structure of DNA.

3 What base is complementary to:
- **a** adenine?
- **b** cytosine?
- **c** guanine?
- **d** thymine?

4 Describe the function of:
- **a** restriction enzymes
- **b** DNA ligase.

5 What vector is used in the production of insulin by recombinant DNA technology?

6 Explain how recombinant DNA technology is used to produce a vaccine for hepatitis B.

APPLY KNOWLEDGE

7 Use a Venn diagram or table to compare and contrast artificial breeding with genetic engineering.

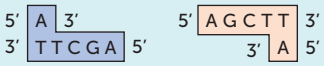
8 Explain why it is possible for an organism of one species to use a gene from another species to produce a protein.

9 Explain the importance of complementary bases with respect to inserting a fragment of DNA into a vector.

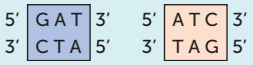
10 What restriction enzyme is the third one isolated from the d strain of *Haemophilus influenzae*?

11 Classify each of the following as blunt ends or sticky ends.

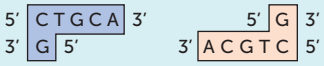
a



b



c



8.2 SYNTHETIC HORMONES

In Chapters 5 and 6, you looked at the important role that hormones play in homeostasis. Unfortunately, some disorders lead to the body being unable to produce certain hormones, which will have significant consequences on the body. A few examples of these disorders are Type 1 diabetes mellitus, hyperthyroidism and hypothyroidism.

Diabetes mellitus

Diabetes, or more correctly **diabetes mellitus**, is a hormonal problem that seriously disrupts homeostasis. A person with diabetes has an abnormally high blood glucose level, a condition called **hyperglycaemia**. As you learnt in Chapter 5, a balance between the hormones insulin and glucagon usually keeps the blood glucose at the correct level for normal body functioning. However, this is not possible in someone with diabetes.

The main role of insulin is to lower the levels of glucose in the blood by stimulating cells to take in glucose, and by liver and muscle cells converting glucose into glycogen. If a person produces insufficient insulin, or if their cells are resistant to the effects of insulin, the amount of glucose in the blood remains high and they excrete large quantities in the urine.

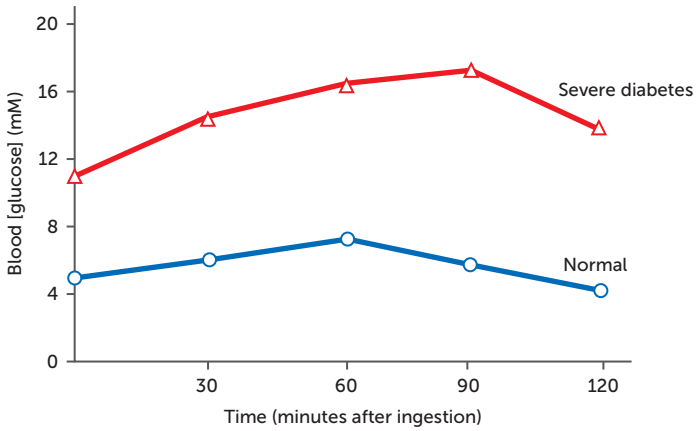


FIGURE 8.12 Glucose levels following ingesting glucose

There are two forms of diabetes: type 1 and type 2.

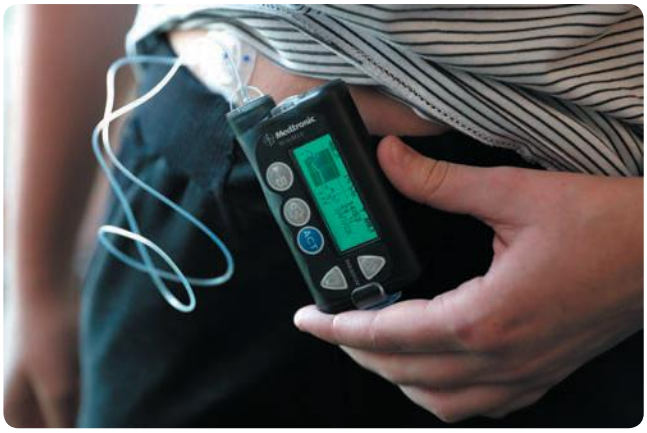
Type 1 diabetes

Type 1 diabetes, sometimes called **insulin-dependent diabetes**, usually begins in childhood, and therefore used to be called juvenile diabetes. In Australia, 10–15% of diabetes patients suffer from type 1. It occurs because a fault in the patient's immune system causes the destruction of beta cells in the islets of Langerhans of the pancreas. Beta cells produce insulin; therefore, a person with type 1 diabetes does not produce insulin. In most cases, the person's cells respond to insulin in the normal way, so the disease can be managed by giving the person insulin.

Insulin cannot be taken in tablet form because it is digested in the alimentary canal. Hence, the only treatment is regular injections of insulin or the use of a programmable pump that provides a continuous supply of insulin under the skin. Insulin injections do not cure type 1 diabetes; they simply fulfil a role to ensure the body is able to function. The patient must have regular injections to stay alive, but even with injected insulin the long-term effects are likely to be kidney failure, heart attack, stroke, amputations, blindness or nerve damage.

Science Photo Library/Michael Donne

FIGURE 8.13 A person suffering from type 1 diabetes is injecting herself with insulin using a NovoPen®, a device that measures the correct insulin dose from a portable cartridge



Alamy Stock Photo/ITAR-TASS News Agency

FIGURE 8.14 An insulin pump. The pump delivers a constant, small dose of insulin with a larger dose before meals or whenever the blood glucose level needs correcting

Type 2 diabetes

Type 2 diabetes is also known as non-insulin-dependent or **adult-onset diabetes**. It usually develops in people over the age of about 45 years, although increasing numbers of younger people are now being diagnosed. Unlike people with type 1 diabetes, type 2 patients are able to produce insulin, but their cells do not respond to it.

Type 2 diabetes is a lifestyle disease; it is more common in people who are not physically active and are overweight or obese. The incidence of type 2 diabetes in Australia and other affluent countries is increasing rapidly due to the large number of people who do not adopt a healthy lifestyle. There are so many Australians developing type 2 diabetes that it has become a health crisis.

Lifestyle factors that increase the risk of developing type 2 diabetes include:

- lack of physical activity
- being overweight or obese
- a diet that is regularly high in fat, sugar and salt, and low in fibre
- high blood pressure
- high blood cholesterol
- smoking.

Type 2 diabetes develops gradually and often there are no symptoms, or they are not noticed. It is estimated that about half of those Australians who have type 2 diabetes have not yet been diagnosed.

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Because the cells do not respond to insulin, they do not take up glucose from the blood. Therefore, a blood test taken after fasting (not eating) will detect abnormally high levels of glucose.

There is no cure for type 2 diabetes, but the earlier a diagnosis is made, the better the chances of successful management of the condition. If it remains undiagnosed or untreated, there is an increasing risk of complications such as heart disease, stroke, kidney disease, eye problems, nerve damage, and skin and foot problems.

The treatment of type 2 diabetes involves a management program that aims to keep blood glucose levels within the normal range. Management includes a careful diet, regular physical activity, maintaining a healthy weight, monitoring blood glucose, and sometimes medication if blood glucose cannot be controlled by other measures.

Type 2 diabetes is preventable. The chances of suffering from the disease can be reduced by adopting a healthy lifestyle.

Diabetes
The Diabetes Australia website provides more information on diabetes, as well as links to many other useful sites including state-based organisations.

Insulin animation
This site contains an animation that explains how insulin is produced.

Treating diabetes

Type 1 diabetes, and sometimes type 2 diabetes, is treated by injections of insulin. The insulin for treatment of diabetics used to be obtained from the pancreases of cows and pigs. This made supplies of insulin expensive and limited. The extracts had to be purified, and patients sometimes suffered allergic reactions or infections from the animal-derived insulin.

In the 1980s, genetically engineered human insulin began to be produced. The gene for human insulin was inserted into DNA of the bacterium *Escherichia coli*. The bacteria were cultured, and the transgene allowed bacterial cells to produce the protein of human insulin. This was then extracted and used to treat people.

Activity 8.3
Investigating the regulation of blood sugar

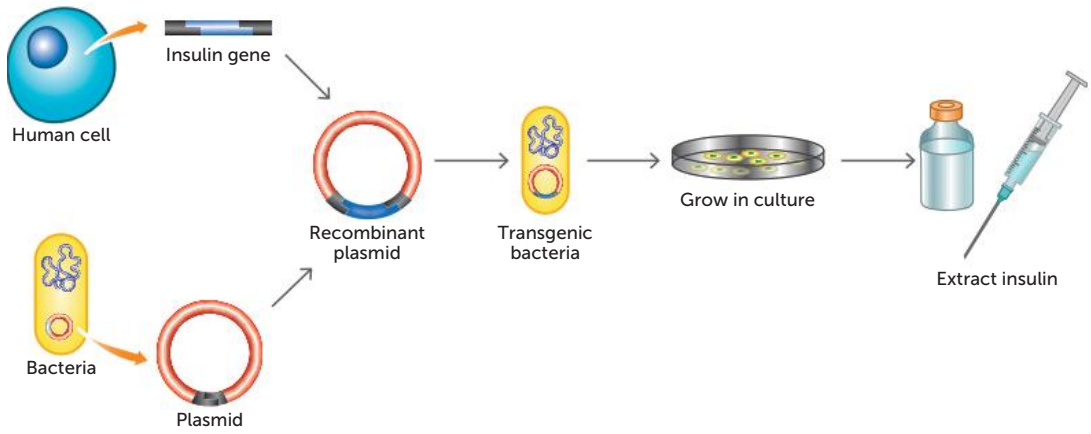

FIGURE 8.15
Recombinant DNA allows bacteria to produce human insulin

8.1 Disruptions to homeostasis

This insulin, produced from recombinant DNA, was marketed as Humulin®. Yeast is now used in a similar way to make insulin, and so almost all the insulin used throughout the world is now biosynthetic recombinant 'human' insulin rather than animal insulin.

Alamy Stock Photo/Science Photo Library

FIGURE 8.16
Scientists working in a facility for the production of recombinant human insulin from the fermentation of yeast cells in these large vats

Thyroid disorders

The thyroid gland is located in the neck and secretes two hormones: **thyroxine (T4)** and **tri-iodothyronine (T3)**. Both have the same effect, but the most important form is thyroxine. Thyroxine affects nearly every tissue in the body by stimulating carbohydrate, protein and fat metabolism. Thus, the secretion of thyroxine from the thyroid regulates basal metabolic rate.

Some of the energy released from the chemical reactions stimulated by thyroxine is in the form of heat, which is important in maintaining body temperature. Thyroid hormones are therefore also important in the long-term homeostasis of body temperature by the gradual change in metabolic rate that occurs from summer to winter.

Secretion of thyroxine is controlled by thyroid-stimulating hormone (TSH). TSH is secreted by the anterior lobe of the pituitary, but its release is controlled by the hypothalamus in the brain.

An excess of or a deficiency in thyroxine can cause disorders. This may be due to a problem in the thyroid itself, but in some cases it can be due to an imbalance in TSH.

Hyperthyroidism

Too much thyroxine, called **hyperthyroidism**, occurs when the thyroid gland produces too much hormone. The most common type of hyperthyroidism is known as **Graves' disease**. It is an enlargement of the thyroid caused by an immune system reaction. Although not inherited, there does seem to be a genetic predisposition for the condition. Because the cells are overstimulated, and the metabolic rate is increased, the symptoms of hyperthyroidism are rapid heartbeat, weight loss, increased appetite, fatigue, sweating, anxiety and, in the case of Graves' disease, protruding eyeballs, known as exophthalmia.

During the production of thyroxine and tri-iodothyronine, iodine is absorbed from the bloodstream, concentrated in cells in the thyroid and then incorporated into the molecules to produce the hormones. Therefore, hyperthyroidism can be treated with drugs that block the thyroid gland's use of iodine. Another method is to give the patient a drink containing radioactive iodine. The radioactive iodine molecules are taken up by the thyroid cells, which are then killed by the radioactivity. Cells elsewhere in the body do not absorb iodine and are unaffected. The radioactive iodine is eventually excreted in the urine. A third method is to use surgery to remove some, or all of, the gland. Less thyroid gland will result in less hormone being produced.

When cells in the thyroid gland are destroyed, there is the risk that the individual may develop hypothyroidism and require treatment with synthetic thyroxine. This is discussed below.

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FIGURE 8.17 Exophthalmia, protruding eyeballs, is a symptom of Graves' disease, the most common result of an overactive thyroid gland

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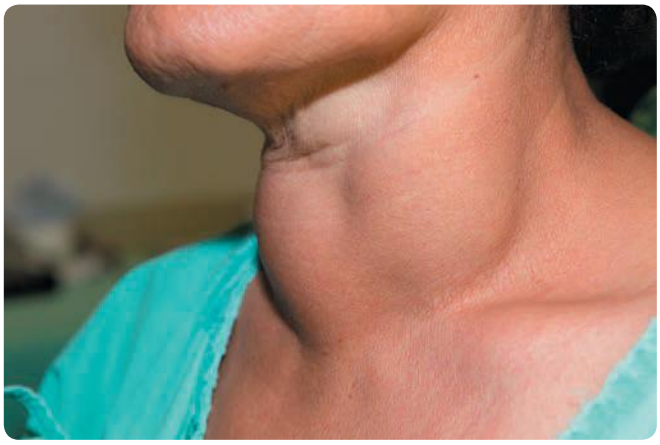
Hypothyroidism

Too little thyroxine, **hypothyroidism**, is much more common than hyperthyroidism. About 6–10% of Australian women, and a smaller proportion of men, may be affected. Hypothyroidism occurs either through problems with the thyroid, pituitary gland or hypothalamus. Symptoms of hypothyroidism arise due to a decrease in metabolism and may include slow heart rate, unexplained weight gain, fatigue or a feeling of lack of energy, intolerance to cold, swelling of the face, and goitre.



Shutterstock.com/Karan Bunjean

FIGURE 8.18 Goitre is the enlargement of the thyroid gland

One thyroid gland problem is due to lack of iodine. A thyroxine molecule contains four iodine atoms (hence T4) and a tri-iodothyronine molecule contains three atoms of iodine (T3). Thus, a deficiency of iodine in the diet can prevent the thyroid gland from making enough hormones. The thyroid gland may then become enlarged in an effort to increase hormone production. Enlargement of the thyroid is known as **goitre**.

Many people may suffer from iodine deficiency without it being severe enough to produce visible swelling of the neck. In Australia, about 46% of people are affected, so iodine deficiency is now a public health problem. To try to ensure that people get sufficient iodine, the federal government introduced compulsory addition of iodine into most breads in October 2009. All bread, except organic bread and bread mixes for baking at home, must now be made with iodised salt.

www.dlibrary.com.au / Saxa

FIGURE 8.19 Using iodised table salt during cooking and with meals can reduce the risk of suffering from hypothyroidism due to iodine deficiency

Although a severe deficiency of iodine can cause hypothyroidism, the most common cause is an attack on the thyroid gland by the patient's immune system. This is known as Hashimoto's disease. Another cause is surgery for cancer of the thyroid that involves removal of all, or a large part, of the gland.

If the cause of hypothyroidism is a lack of iodine, it is easily treated by the inclusion of extra iodine in the diet. For treatment of other causes, tablets containing thyroid hormone are prescribed. There is no cure and the hormone tablets must be taken for the rest of the person's life. The dose of thyroid hormone must be carefully monitored because too little will not relieve the symptoms of hypothyroidism but too much will result in hyperthyroidism.

Hypothyroid patients used to be treated with tablets made from the dried and powdered thyroid glands of animals, mainly pigs. The tablets contain both thyroid hormones T3 and T4 but not necessarily in the same proportions as produced by the human thyroid. They also contain traces of other hormones. Although these 'natural' tablets are still available, today most patients are treated with hormones made synthetically by a chemical process.

Thyroxine was first isolated in 1914 and was synthesised for the first time in 1927. Levothyroxine is a manufactured form of thyroxine, and is now considered so safe and effective that it is listed on the World Health Organization's List of Essential Medicines. It is available in both oral and injectable forms under many different brand names. In Australia, levothyroxine is sold as Oroxine, Eutroxsig or Eltroxin. Levothyroxine is the most commonly prescribed drug for thyroid hormone replacement.

Thyroid gland
The Thyroid Australia website provides more information about the thyroid and links to relevant websites.

Activity 8.4
Investigating thyroid hormone

Key concept

Synthetic hormones produced by either recombinant DNA or chemical manufacturing can be used to treat disorders such as diabetes mellitus and hypothyroidism.

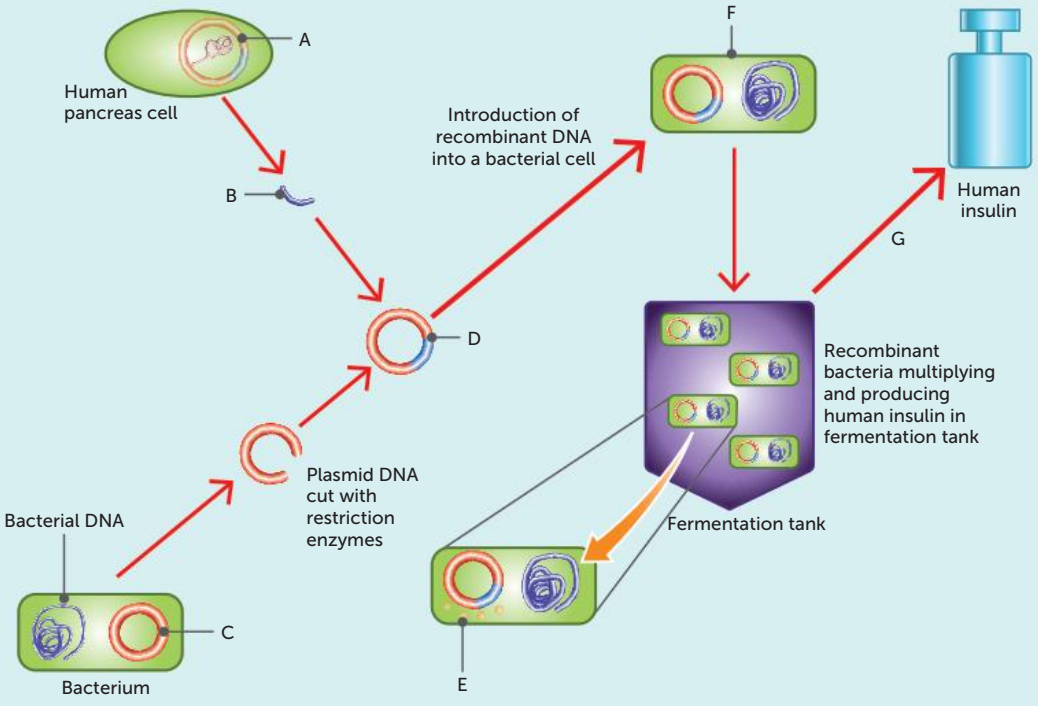
Questions 8.2

RECALL KNOWLEDGE

1 Define 'hyperglycaemia' and explain why a lack of insulin results in this condition.
2 What causes type 1 diabetes?
3 List the symptoms of hyperthyroidism.
4 a Is goitre a symptom of hypothyroidism or hyperthyroidism?
  b Explain why goitre occurs.
5 Explain why iodine supplements are used to treat some forms of hypothyroidism.
6 Explain why levothyroxine is considered the preferred treatment for hypothyroidism.

APPLY KNOWLEDGE

7 Explain why insulin in not an effective treatment for type 2 diabetes.
8 Insulin pumps are programmed to deliver a surge of insulin after meals and a small, steady rate of insulin at other times. Explain why this is preferred over traditional injections.
9 Suggest why type 2 diabetes is more common in adults.
10 Label parts A–G on the diagram below to show the steps involved in producing insulin.



11 Thyroxine hormone replacement is used to treat hypothyroidism. Explain why some patients with hyperthyroidism may need to receive hormone replacement after initial treatments.

8.3 OTHER TECHNOLOGIES

Gene therapy and cell replacement therapies are also used to treat disorders.

Gene therapy

Gene therapy aims to treat or cure genetic abnormalities by identifying faulty genes and inserting healthy ones. It is a way of using the genes themselves as the treatment. In many ways, it is the most obvious application of the **Human Genome Project**, which has revealed the location of around 4000 potentially faulty genes. Currently, gene therapy research is concentrating on single-gene disorders such as cystic fibrosis, Huntington's disease, muscular dystrophy and sickle-cell anaemia. It is also being investigated for curing type 1 diabetes. Unlike most conventional medicines, which treat the symptoms of a disease, gene therapy has the potential to correct the underlying cause.

Gene therapy was first introduced in 1970. However, it is still only at a research level. Some of the areas of possibility are:

- replacing a mutated gene with a healthy copy
- fixing or inactivating mutated genes
- inserting a new gene that will fight the disease
- making the immune system recognise diseased cells.

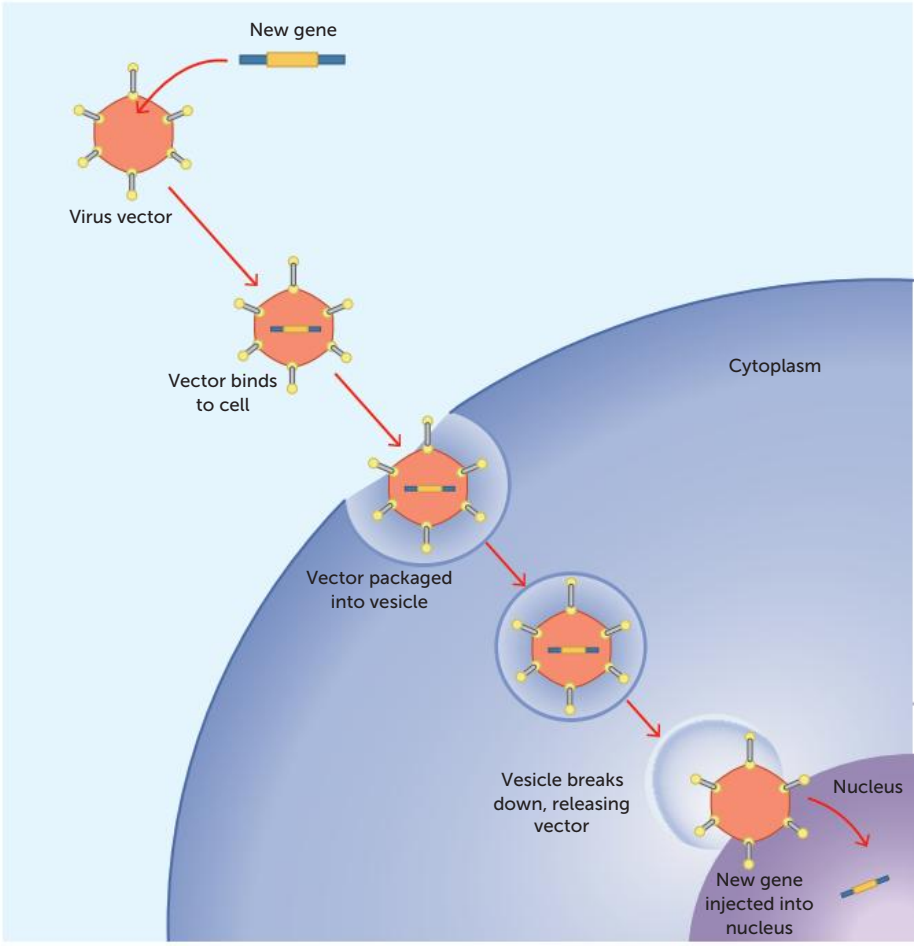


FIGURE 8.20 A gene being introduced into a cell during gene therapy

The concept of gene therapy is that a vector can be used to deliver desired DNA into a cell. This DNA can be incorporated into the cell's nucleus and undergo transcription and translation to produce the desired protein.

Key concept

Gene therapy aims to correct the cause of the problem by correcting the faulty gene. This can be achieved by replacing, correcting or inactivating the gene, or by inserting a new gene to correct the disease or to encourage the immune system to destroy the diseased cell.

Sickle-cell anaemia
This website provides information about recent progress with gene therapy for sickle-cell anaemia.

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Type 1 diabetes

Earlier in this chapter, you saw how type 1 diabetes is caused by an autoimmune disease that destroys the beta cells in islets of Langerhans in the pancreas. This means that the body cannot produce insulin and, therefore, blood glucose levels remain high after a meal.

Traditional treatment of diabetes focuses on introducing the insulin that the body cannot make. However, gene therapy is now looking at methods of making it possible for the body to produce insulin again.

Gene therapy
This article discusses how gene therapy could be used to cure type 1 diabetes.

One possibility is reprogramming other cells to produce insulin. In order to achieve this, the gene for insulin is introduced into a vector. The vector is then used to 'infect' the desired cells, such as the alpha cells in the islets of Langerhans. These cells incorporate the new DNA into their nucleus and are able to use protein synthesis to produce insulin.

Key concept

Type 1 diabetes could possibly be treated with gene therapy by using vectors to introduce the gene for insulin into alpha cells so that they can produce the hormone and allow the body to function normally.

Cystic fibrosis

Cystic fibrosis (CF) is the most common life-threatening genetic disorder among Australians of European descent. It mainly affects the lungs and pancreas, but sometimes the liver and reproductive organs. CF is characterised by thick sticky mucus secreted by the mucous glands. In the lungs, this mucus may clog the tiny air passages and trap bacteria, making a person with CF susceptible to infection. Repeated infections and continual blockage of the airways may cause irreversible lung damage and shorten life expectancy. The pancreas is also affected, preventing secretion of enzymes required for digestion. Therefore, people with CF frequently have problems with nutrition and need to take care with their diet.

CF results when an individual inherits the recessive allele for the condition from each parent. In most Australian states, a blood sample is usually taken from a baby's heel within two to three days after birth. If the blood test reveals that a child has the disease, a special low-fat, high-carbohydrate and high-protein diet is advised.

The identification of the Cystic Fibrosis Transmembrane Regulator (CFTR) gene in 1989 was a major step forward in developing a treatment for CF. Mutations in this single gene result in the disease, and since its discovery more than 900 mutations have been identified. In 1991, scientists successfully corrected faulty CFTR genes in cultured cells by adding normal copies of the gene to the culture. This was the first step towards gene therapy for CF.

CF was a logical choice for treatment using gene therapy. It is a single-gene disorder, and the most severely affected organ, the lung, is relatively easy to access to provide treatment. In addition, the disease is slow to progress, with the lungs of a newborn being virtually normal. This would enable gene therapy to begin before significant lung damage started to occur. The first experimental gene therapy treatment was given to a patient with CF in 1993. Researchers modified a common cold virus to act as the vector to carry normal genes to the CFTR cells in the airways of the lung. This first study was mainly concerned with the safety issues of the treatment. The amount of gene transfer was probably too small to have any real therapeutic benefit and any benefit was short-lived. Trials with alternative methods of gene transfer are continuing.

Huntington's disease

Another single-gene disorder is **Huntington's disease**, and researchers believe that gene therapy could be used to slow down or prevent its development. It is caused by a mutation in a single gene on chromosome 4 called IT15. The symptoms of this incurable genetic disorder seldom appear before the age of 40. The mutated form of a protein called huntingtin results in nerve cells in the brain

being damaged, causing physical, mental and emotional changes. The disease is characterised by occasional unintentional flailing movements of the arms and legs, and difficulty in making voluntary movements of the limbs. The affected person also suffers from progressive dementia, the loss of ability to think clearly.

Research in the United States on mice has indicated that gene therapy for Huntington's disease could be effective in humans. In other research, French scientists experimented with a modified virus to deliver a corrective gene into brain cells that boosts a natural shield against the effects of the defective huntingtin protein. This research has been conducted on rats and primates, and the positive results have encouraged movement towards a clinical trial on humans.

Key concept

Genetic disorders such as cystic fibrosis and Huntington's disease could possibly be treated with gene therapy by introducing the correct gene into the affected cells so that they can function normally.

Cell replacement therapy and tissue engineering

Stem cells are undifferentiated cells that are capable of repeated mitotic divisions for long periods of time and, given the right conditions, can differentiate into specialised cells. These characteristics make them ideal for producing replacement tissues. In *Human Perspectives ATAR Units 1 & 2*, the types of stem cells and their sources were discussed.

Any disorder involving loss of, or injury to, normal cells is a potential candidate for stem **cell replacement therapy**. However, cell replacement therapy for the nervous system has generated the most interest, due to the debilitating nature and widespread occurrence of neurodegenerative disorders such as Parkinson's and Alzheimer's diseases. The most attractive method for restoring brain function in, say, Parkinson's disease is the replacement of dying neurons with healthy neuronal tissue. Pilot studies using embryonic stem cells have been carried out in humans with some success. The transplanted cells not only survived but also grew and established connections with adjacent neurons. However, the use of human embryonic stem cells is controversial and raises a number of ethical questions. Researchers into Parkinson's disease are currently exploring other sources of cells to help restore patients' brain function.

Stem cells are increasingly being used for tissue engineering. The primary objective of **tissue engineering** is to restore healthy tissues or organs for patients and thus eliminate the need for tissue or organ transplants, or artificial implants. Early research in this area used cells from the intended recipient but, in many cases, such as genetic diseases, this was not practicable. In other situations, the organ from which cells were to be harvested was diseased, and so not enough normal cells were present to enable a successful culture. The use of stem cells overcomes both problems.

Tissue engineering
This website provides an interesting account of how tissue engineering is being used to manufacture artificial skin.

Tissue engineering requires an abundant supply of disease-free cells of specific types. These cells then need to be induced to grow on a **scaffold** of natural or synthetic material to produce a three-dimensional tissue. Tissue engineering scaffolds serve as a template for tissue growth, and need to have high pore sizes that enable the cells to grow while at the same time allowing the diffusion of nutrients throughout the whole structure. They frequently need to be biodegradable so that they can be absorbed by the surrounding tissues without having to be removed surgically. This needs to be carefully established, as the rate at which the scaffold degrades needs to match, as far as possible, the rate of tissue formation. That is, while the new cells are manufacturing their own natural matrix structure around themselves, the scaffold is providing a support structure that will eventually break down, leaving newly formed tissue.

Once a scaffold has been devised, suitable stem cells need to be cultured. These cells are seeded on to the scaffold, which then enables further cell growth and proliferation. This cell-covered scaffold is then implanted into the patient at the site where new tissue is required. As the new cells continue to grow and divide, the material making up the scaffold begins to degrade or, in some cases, to be absorbed. Such tissue engineering techniques are being used to develop a wide range of tissues, including bone, skin, cartilage and adipose tissues.

Key concept

Stem cells can be utilised in cell replacement therapy and tissue engineering due to their ability to multiply and differentiate.

Questions 8.3

RECALL KNOWLEDGE

1. Define 'gene therapy', and give an example of its possible use.
2. List three ways that gene therapy could possibly correct faulty genes.
3. Explain the role of a vector in gene therapy.
4. Explain why it is important to diagnose cystic fibrosis at infancy.
5. Huntington's disease results in the death of brain cells. Explain how scientists believe gene therapy could work to treat this disease.
6. What are stem cells, and why are they suitable for cell replacement therapy?
7. Why is it possible that cell replacement therapy could be used to treat patients with Alzheimer's or Parkinson's disease?

APPLY KNOWLEDGE

8. Suggest why gene therapy is more difficult than recombinant DNA technology to develop safely and effectively.
9. Discuss why gene therapy for type 1 diabetes is focused on introducing the gene into alpha cells.
10. Discuss why research into cell replacement therapy has focused on neurological conditions.
11. Tissue engineering utilises a scaffold for the tissues to grow on. Explain why this is necessary, and why it needs to be biodegradable.
12. 'Gene therapy has suffered from skepticism from both [the] scientific community and [the] pharmaceutical industry. In addition to the risk of insertional mutagenesis/tumorigenesis, the widespread clinical application of gene therapy is hampered due to the inefficient systemic delivery. However, in recent years, new approaches, including stem cell-based gene therapy, have boosted the potential comeback of gene therapy' (Ye, Z and Mahato, R, 'Combining Stem Cells and Genes for Effective Therapeutics', NCBI Online, 2009: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3207237/ [Accessed 30 August 2020].
    a. Suggest how stem cell replacement therapy can be combined with gene therapy.
    b. Discuss why tumour growth is a possible risk with this technology.
    c. Suggest a disorder for which this combined therapy may be a viable treatment, and explain how it could work.

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CHAPTER 8 ACTIVITIES

ACTIVITY 8.1 Investigating restriction enzymes

Recombinant DNA technology, or genetic engineering as it is frequently called, involves the introduction into cells of fragments of DNA that are foreign to the organism. To do so, the strands of DNA under investigation need to be cut into useful fragments. The 'scissors' that cut the DNA are called restriction enzymes. The fragments can then be inserted into a suitable vector and joined with DNA ligase.

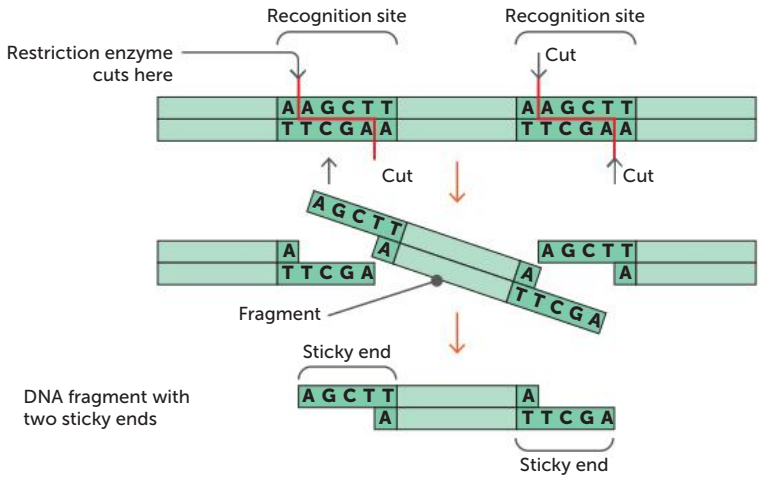
In this activity, we will investigate how a sequence of DNA can be cut into suitable fragments using an appropriate restriction enzyme.

What to do

Answer the questions below, referring to the relevant parts of this chapter where necessary.

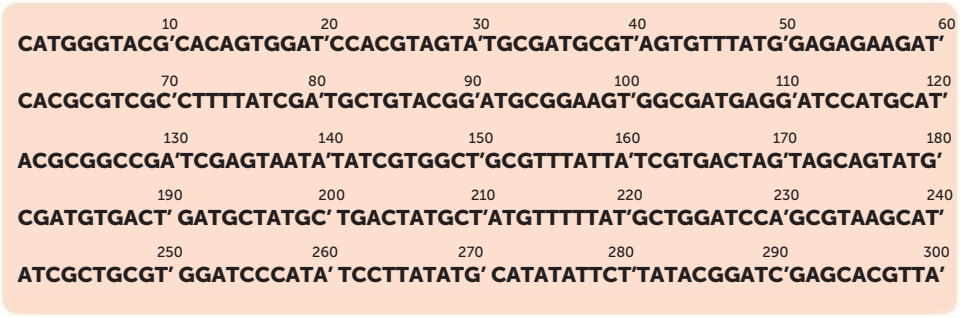
1 Explain the following terms by describing their role in recombinant DNA technology.
   - **a** Restriction enzymes
   - **b** Recognition sites
   - **c** Blunt ends
   - **d** Sticky ends

2 Use Table 8.1 (page 203) to identify the restriction enzyme that is being used in the following figure and the organism from which it was first isolated.



A restriction enzyme cuts a double-stranded DNA molecule at the recognition site

3 Imagine that you are a genetic engineer and need to cut the DNA sequence shown on the following page. Using the five restriction enzymes listed in Table 8.1, study the sequence carefully and circle every recognition site that could be cut by each of the enzymes in turn. You may wish to use pens or pencils of four different colours. How many fragments of DNA have you created for each enzyme?



A single strand of DNA

4 A process called ligation is used to reassemble the fragments. Name the enzyme involved in this process.
5 Explain why the process of ligation can be viewed as the reverse of the restriction enzyme procedure.
6 Use a short summarising statement to explain why the discovery of restriction enzymes and DNA ligase has been so important for the advancement of genetic engineering.

SOUTHERN Biological Developed by Southern Biological

ACTIVITY 8.2 Investigating bacterial transformation

DNA can mutate spontaneously or after an error is made in DNA replication. Biotechnologists have developed methods of controlled DNA mutation, such as intentionally mutating cell DNA to alter how the cell behaves. However, it is also possible to transfer DNA from one organism into another. This method, called genetic transformation, uses an engineered molecule of DNA to transfer a gene or genes from one organism to another so that the organism is capable of producing the protein encoded by the transformed gene.

Aim

To perform bacterial transformations using the green fluorescent protein plasmid pGreen
Time requirement: 50 minutes

You will need

Escherichia coli (E. coli) MM294 starter plate; 10 µL pGreen plasmid; 2 Luria broth agar plates; 2 Luria broth with ampicillin agar plates; 500 µL Luria broth, sterile; 500 µL calcium chloride ($CaCl_2$), 50 mM, sterile; 2 transformation tubes, sterile; 8 plastic pipettes, 1 mL, sterile; 3 inoculation loops, sterile, disposable; 4 inoculation spreaders, sterile, disposable; 2–20 µL variable micropipette; sterile tips for 2–20 µl micropipette; water bath; thermometer (if necessary); ice bath; fine-point marker pen; stopwatch; microtube rack; sticky tape (to seal plates); incubator; ethanol (or bleach); disposable gloves

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Some bacteria may cause disease, so assume them to be pathogenic. (Note: *E. coli* MM294 is a harmless school-safe biological.)	Wear lab coats, safety glasses and gloves; wash hands thoroughly at end. Decontaminate benches before and after activity. Flood spills with bleach.
Micro-organisms will grow on the agar plates.	Do not open plates once they are securely taped. Dispose of plates appropriately after autoclaving.
Disinfectants or bleach may leave a corrosive residue.	After wiping the bench clean with bleach, ensure the residue is wiped off; ensure lab coat sleeves are rolled down and gloves are worn.

What to do

Note: To use aseptic technique, wipe your bench down with ethanol (or bleach) and keep your work near the Bunsen burner to waft potential contaminants away from your materials.

Preparing the transformation solution

1. Label one transformation tube '+ Plasmid' and the other '– Plasmid'. Keep the tubes cold by placing them upright in the ice bath. Keep tubes capped at all times except when in use.
2. Add 250 µL (0.25 mL) of ice-cold calcium chloride $CaCl_2$ solution to each transformation tube, using a sterile plastic pipette. Maintain the temperature by placing the tubes back into the ice bath.

Suspending the bacteria

1. Transfer a single colony of *E. coli* from the starter plate to the ice-cold $CaCl_2$ solution in the '+ Plasmid' transformation tube using a sterile inoculation loop. To dislodge the *E. coli* cells from the loop, spin the loop rapidly in the solution. Check whether your *E. coli* has transferred successfully – it should be visible in your tube.
2. Suspend the *E. coli* in the $CaCl_2$ solution by drawing the solution in and out of a sterile pipette by squeezing and releasing the bulb several times. You should see the solution begin to become milky white as cell mass is suspended. To check there are no lumps or particles in the tube, hold it up to the light; then return the tube to the ice.
3. Repeat the same steps to transfer a single colony of *E. coli* from the starter plate to the ice-cold $CaCl_2$ solution in the '– Plasmid' transformation tube.

Adding the plasmid

1. The technician/teacher will bring the plasmid to your workstation. Transfer 10 µL (0.01 mL) of plasmid solution to the transformation tube labelled '+ Plasmid' using a micropipette. Add the plasmid directly to the liquid in the tube without allowing it to touch the sides.
2. Immediately return the tube to the ice bath and mix the plasmid into the bacterial suspension by placing a sterile inoculation loop into the liquid and rapidly spinning it with your fingers. Cap the tube when done. Incubate the two tubes for 15 minutes on ice.
3. Label the four plates as follows:
   - LB + Plasmid
   - LB – Plasmid
   - LB/Amp + Plasmid
   - LB/Amp – Plasmid

Heat shock

1. Once the 15-minute incubation is finished, take the two tubes from the ice bath and transfer to the warm-water bath (42°C) and hold them for 90 seconds with your gloved hands, keeping the tube caps from being fully submerged in the water. Gently agitate the tubes while they are warming up in the water. Immediately move the tubes back to the ice bath when the time is up.
2. Allow the tubes to rest in the ice bath for at least 1 minute before continuing.

Recovery

1. Add 250 µL (0.25 mL) of Luria broth to each tube using a sterile plastic pipette. Hold the capped tubes at the top and gently tap/flick the base with your finger to mix the liquids.
2. Allow tubes to recover for 10 minutes in a microtube rack at room temperature.

Plate inoculation

1. Transfer two drops of liquid from the '+ Plasmid' tube to the 'LB + Plasmid' plate using a sterile plastic pipette. Quickly spread the liquid evenly over the plate surface using a sterile spreader.
2. Repeat for the 'LB/Amp + Plasmid' plate.
3. Transfer two drops of liquid from the '– Plasmid' tube to the 'LB – Plasmid' plate using a sterile plastic pipette. Quickly spread the liquid evenly over the plate surface using a sterile spreader.
4. Repeat for the 'LB/Amp – Plasmid' plate.

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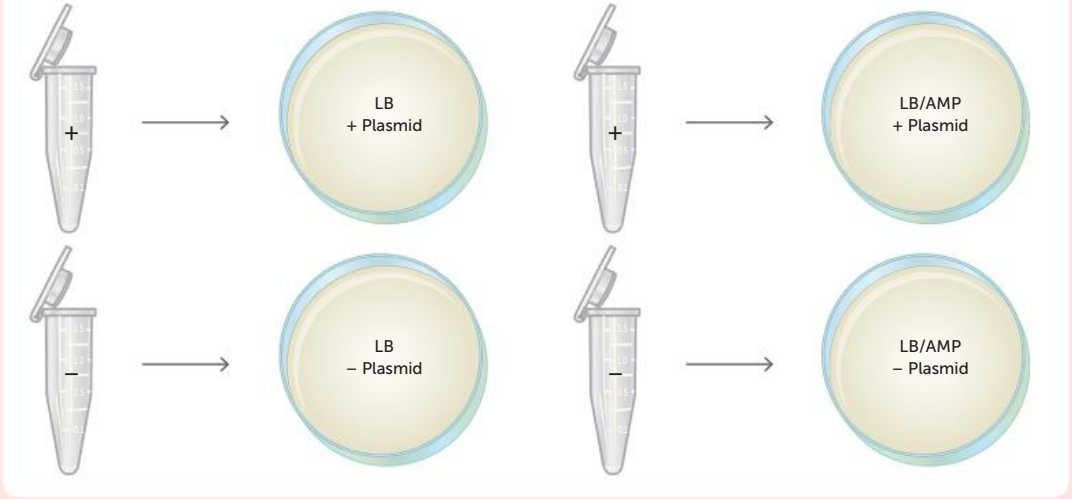

5 Secure the lid of each Petri dish to its base using sticky tape. Leave plates to rest on the bench for 5 minutes and then place them upside down (agar on top) in a 33°C incubator for 24–36 hours. You can inspect growth after this time. You should see either a bacterial lawn (colonies of bacteria covering all or most of the plate), single colonies (spots), or no growth on the individual plates. Take the plates into a dark room to observe evidence of fluorescence in the transformed colonies. Use of a UV light may enhance the fluorescence.

6 To count the number of individual colonies, mark the lid of the Petri dish above each colony with a marker as you count it. Mark any plates with cell growth too dense to count as individual colonies, as a lawn. Record your results.

Studying your results

1 Record the results of your experiment by copying the table below.

Bacteria colony results

PLATE	RESULT
– Plasmid on LB agar	
+ Plasmid on LB agar	
– Plasmid on LB/Amp agar	
+ Plasmid on LB/Amp agar	

2 What growth and phenotypes can you observe?

3 Describe what you see on your plates when you look at your plates under UV light.

Discussion

1 Explain what a plasmid is.

2 Why is the plasmid–bacteria solution placed on ice for 5 minutes?

3 Which plate forms the control in this experiment? Explain.

4 Explain the function of the LB broth. What is the purpose of incubating the cells at room temperature?

5 Explain how the DNA plasmid is put into bacteria. What is the advantage of being able to do this? Consider what the plasmid DNA allows the bacteria to do.

6 Explain how we are able to identify that the plasmid DNA is in the bacteria.

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ACTIVITY 8.3 Investigating the regulation of blood sugar

1 Two men (A and B) were subject to a glucose tolerance test. Each was given 100 g of glucose at the start of the experiment. The table below shows their blood glucose concentration during the period of the experiment.

TIME SINCE START (HOURS)	MAN A(mg/100 mL)	MAN B (mg/100 mL)
0	80	170
0.5	100	250
1	160	310
1.5	130	300
2	80	280
3	60	210
4	55	180
5	90	160

a Plot the data for the two men as a graph.
b One of the men had a diseased pancreas. Which man was it? Give reasons to support your answer.
c What is the name of the disease from which the man was suffering?

2 Research workers who were investigating blood glucose regulation injected hormones singly, or as mixtures, into the vein of a dog for five hours. They then measured any increase in blood glucose above the normal level. Their results are shown in this table.

HORMONE INJECTED	RISE IN BLOOD GLUCOSE LEVEL (mg/100 mL)
Adrenaline	30
Glucagon	10
Cortisol	3
Adrenaline and glucagon	58
Adrenaline and cortisol	58
Glucagon and cortisol	35
Adrenaline, glucagon and cortisol	140

a Which parts of the endocrine system are responsible for releasing each of the hormones listed in the table?
b A technician suggested giving the hormones to the dog by adding them to its drinking water. Would this method be effective? Explain.
c Compare the effects of these hormones when acting:
i singly
ii together.
d What do the data indicate about the response of body tissues to the hormone mixtures?
e In light of your answers to parts **c** and **d**, explain how stress could raise blood glucose levels. What would be the advantage of this response?
f Describe at least three major criticisms of the design of the experiment on the dog.

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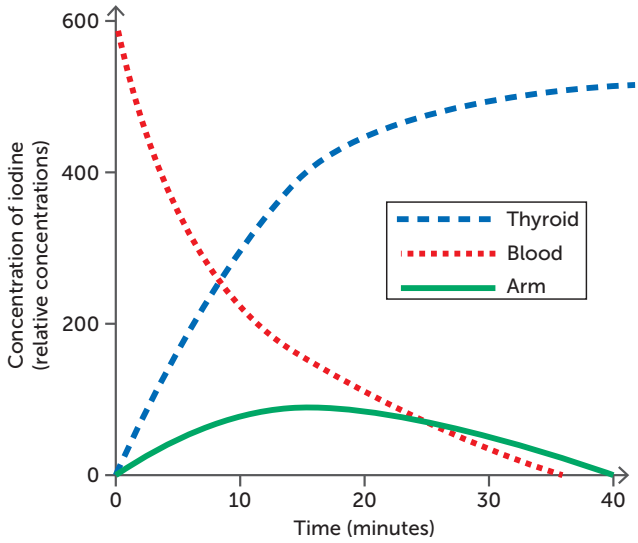
ACTIVITY 8.4 Investigating thyroid hormone

A scientist injected radioactive iodine into a blood vessel in a person's arm. He then measured the concentration of iodine in the arm, in the blood and in the thyroid gland for the next 40 minutes.

The following graph shows the concentrations of iodine measured by the scientist.



Graph showing iodine concentrations in the thyroid and blood over time, after injection into an arm

1. Suggest a hypothesis the scientist may have been testing.
2. Why was the scientist using iodine, rather than some other substance, to investigate the thyroid gland?
3. Why was radioactive iodine used?
4. What do you think the scientist was trying to demonstrate with this experiment?
5. Using the graph, explain what happened to the iodine in the 40 minutes after it was injected into the arm.
6. An important part of the investigation was to measure the concentration of iodine in the person's arm. Why was this necessary?
7. Would it have made any difference to the investigation if the iodine had been injected into the person's leg?

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CHAPTER 8 SUMMARY

- Biotechnology uses cellular processes to make products that are used by humans. It includes newer processes such as recombinant DNA, gene therapy and cell replacement therapy.
- DNA is a molecule made up of nucleotides joined in two strands twisted to make a double helix. Each nucleotide is composed of a sugar, phosphate and nitrogenous base.
- Nitrogenous bases are complementary, and only bond to the complementary pair. Adenine is complementary to thymine, while cytosine is complementary to guanine.
- Recombinant DNA technology is also known as genetic engineering. It involves artificially modifying DNA and it produces genetically modified organisms (GMOs).
- Transgenic organisms are an example of GMOs where DNA from one species is introduced into another species.
- Restriction enzymes are isolated from bacteria. They recognise a certain base sequence, the recognition site, and separate the nucleotides at a certain position. As this occurs on both strands, the DNA is opened with either a straight cut producing blunt ends or a staggered cut with sticky ends.
- Restriction enzymes are named by the bacterium that they are isolated from.
- DNA ligase is an enzyme that joins the phosphate of one nucleotide to the sugar of another. This joins two segments of DNA together.
- Vectors are used to transfer DNA from one organism to another. Restriction enzymes are used to cut the DNA at the same sequence on the vector and the DNA to be inserted. DNA ligase is then used to join the two pieces of DNA together.
- Plasmids are circular pieces of DNA that are often found in bacteria. Bacteriophages (phages) are viruses that infect bacteria. Plasmids and phages are commonly used as vectors.
- Recombinant DNA is used to produce hormones, including insulin, and vaccines such as the hepatitis B and HPV vaccines.
- DNA vaccines are currently being researched. These introduce the DNA for the antigen into the host cells. The host cell can produce the antigen and initiate an immune response.
- Diabetes mellitus leads to hyperglycaemia, as insulin is unable to reduce the blood glucose levels.
- Type 1 diabetes occurs when the immune system destroys beta cells, which means that they cannot produce insulin. It is managed by insulin injections. This insulin can be synthesised by recombinant yeast cells which have had the gene for insulin introduced by a plasmid vector.
- Type 2 diabetes usually develops in adults as a result of lifestyle choices such as obesity and lack of exercise. It is due to the cells being unable to respond to insulin. It is managed by using diet and exercise to keep blood glucose levels within the normal range.
- The thyroid gland produces thyroxine and tri-iodothyronine, which regulate metabolism.
- Hyperthyroidism is when there is too much thyroxine produced, which increases the rate of metabolism. It is treated by blocking the uptake of iodine, using radioactive iodine to destroy thyroid cells, or surgically removing some of the thyroid gland.
  The destruction of thyroid cells can lead to hypothyroidism.
- Hypothyroidism is when there is not enough thyroxine and so the rate of metabolism is too slow. A lack of iodine can lead to hypothyroidism; therefore, iodine supplements can be used as a treatment.
- Another cause of hypothyroidism is an autoimmune disease that destroys the

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thyroid gland. In this case, a synthetic thyroxine called levothyroxine is used as a treatment.

- Gene therapy aims to work by inserting genes to perform the function of abnormal genes. It is currently in the research stage, but is hoped to be used for disorders such as cystic fibrosis, Huntington's disease and type 1 diabetes.
- Gene therapy uses a vector to introduce DNA into the host cells. Research is being conducted into introducing the gene for insulin into alpha cells so that they take on the role of beta cells in the body of people with type 1 diabetes.
- Cystic fibrosis and Huntington's disease are due to a faulty gene. Gene therapy research is looking into using a vector to introduce normal genes into cells to allow them to function normally.
- Stem cells can be used in cell replacement therapy as they have the ability to multiply and differentiate. In particular, this raises the possibility of replacing dying neurons in patients with Alzheimer's or Parkinson's disease.
- Tissue engineering can be used to replace damaged tissue. It is achieved by using a scaffold with stem cells. The scaffold degenerates as the cells multiply, producing new tissue.

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CHAPTER 8 GLOSSARY

Adult-onset diabetes A common form of diabetes that usually occurs in people over the age of 45 who are overweight; it can usually be controlled by diet; also known as type 2 diabetes

Artificial selection An ancient form of genetic engineering where humans select desired traits and choose parents based on these traits

Bacteriophage A virus that infects bacteria

Biotechnology The use of biological processes to produce useful products

Blunt end The end produced by a straight cut of a sequence of nucleotide bases

Cell replacement therapy The replacement of damaged cells with healthy ones

Cystic fibrosis (CF) A disorder controlled by a recessive allele carried on an autosome that is incurable but can be detected during foetal development; mucus-secreting glands, particularly in the lungs and pancreas, become fibrous and produce abnormally thick mucus, resulting in, among other things, chest infections

Diabetes *see* diabetes mellitus

Diabetes mellitus A group of diseases, all of which result in an abnormally high level of glucose in the blood and excretion of glucose in the urine; common name is diabetes

DNA ligase An enzyme capable of combining two small components of single-strand DNA into one single structure

DNA vaccine A vaccine that stimulates an immune response by introducing antigen DNA, which causes the host cells to produce the antigen

Endonuclease An enzyme that breaks a nucleic acid within the strand by separating two nucleotides

Gene therapy The treatment of disease by replacing, manipulating or supplementing non-functional genes in cells and tissues

Genetic engineering *see* recombinant DNA technology

Genetically modified organism (GMO) An organism produced by genetic engineering

Goitre A swelling of the neck caused by an enlargement of the thyroid gland

Graves' disease A medical condition in which overactivity of the thyroid gland results in the secretion of excess amounts of the thyroid hormones

Human Genome Project A project with the aim of mapping the base pairs and identifying the genes in human DNA

Huntington's disease An inherited disease that causes the death of brain cells and results in changes in mood, a lack of coordination and an unsteady gait

Hyperglycaemia An abnormally high level of sugar in the blood; frequently found in people with diabetes mellitus

Hyperthyroidism Overactivity of the thyroid gland resulting in abnormally high levels of thyroid hormones in the blood

Hypothyroidism Underactivity of the thyroid gland resulting in low levels of thyroid hormones in the blood

Insulin-dependent diabetes A form of diabetes that develops rapidly, usually before the age of 20; caused by a decline in insulin-producing cells of the pancreas; treated by injections of insulin at regular intervals; also known as type 1 diabetes

Ligation The process of joining short strands of DNA during replication

Palindromic A sequence that reads the same backwards and forwards

Phage *see* bacteriophage

Plasmid In a bacterial cell, small circular strands of DNA distinct from the main bacterial genome; composed of only a few genes and able to replicate independently within cells

Recognition sequence The sequence of bases in the recognition site

Recognition site A specific sequence of nucleotides at which an enzyme cuts a strand of DNA

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Recombinant DNA Synthetic DNA; made by inserting genes from one source into a DNA molecule from a different source

Recombinant DNA technology The procedures used to produce recombinant DNA; involve introducing DNA into a cell from a different type of organism or DNA that has been modified in some way

Recombinant vaccine A vaccine produced through recombinant DNA technology

Restriction enzyme An enzyme that cuts strands of DNA at a specific sequence of nucleotides

Scaffold A structure used in tissue engineering as a template for tissue growth

Selective breeding *see* artificial selection

Staggered cut A cut produced when a restriction enzyme creates fragments of DNA with unpaired nucleotides that overhang at the break in the strands; called sticky ends

Sticky end The overhanging end produced by a staggered cut of a sequence of nucleotide bases; sometimes called cohesive end

Straight cut A cut produced when a restriction enzyme makes a clean break across the two strands of DNA so that the ends terminate in a base pair; called blunt ends

Thyroxine (T4) A hormone secreted by the thyroid gland that regulates metabolism, growth and development

Tissue engineering The rebuilding of damaged tissue by the use of biology, medicine and engineering

Transgenic organism An organism that has had DNA from another species introduced into it artificially

Tri-iodothyronine (T3) A hormone secreted by the thyroid gland; contains iodine and is the most powerful of the thyroid hormones; affects many body processes, including body temperature, growth and heart rate

Type 1 diabetes *see* insulin-dependent diabetes; also see diabetes mellitus

Type 2 diabetes *see* adult-onset diabetes; also see diabetes mellitus

Vector A bacterial plasmid, viral phage or other such agent used to transfer genetic material from one cell to another

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CHAPTER 8 REVIEW QUESTIONS

Recall

1 a What is recombinant DNA technology?
  b List three possible applications of recombinant DNA technology.
2 a What are restriction enzymes?
  b How are recognition sites related to restriction enzymes?
  c List examples of restriction enzymes. For each, give their bacterial origin.
  d Differentiate between 'sticky' and 'blunt' ends in relation to restriction enzymes.
3 What is DNA ligase, and what is it used for?
4 a What are vectors, and how are they used in recombinant DNA technology?
  b List two different types of vectors that are used in this technology.
5 Which of the two types of diabetes can frequently be treated by modifying the patient's behaviour? Explain the nature of the behaviour modification that is necessary for effective treatment.
6 a What is gene therapy?
  b How is gene therapy likely to advance the treatment of type 1 diabetes, cystic fibrosis and Huntington's disease?
7 a Define 'cell replacement therapy'.
  b How could cell replacement therapy aid the treatment of diseases such as Parkinson's and Alzheimer's?
8 List two diseases that are being prevented with recombinant vaccines.

Explain

9 Explain, with examples, how a transgenic organism is considered a GMO.
10 Explain the differences between type 1 and type 2 diabetes.
11 Explain how the treatment of type 1 diabetes has been assisted by recombinant DNA technology.
12 Explain why scaffolds are used in tissue engineering.
13 Explain the difference between hyperthyroidism and hypothyroidism.
14 Explain how a dietary deficiency can cause hypothyroidism.
15 Annotate a diagram to explain how biosynthetic insulin is produced.
16 Drugs used to treat thyroid deficiency are produced synthetically. What advantages are there in using synthetic drugs rather than those obtained naturally?

Apply

17 Name the third restriction enzyme isolated from *Haemophilus aegyptius*.
18 The diagram below shows the base sequence for a section of DNA.

GGTCAAGCTTACTCGGATCCAGCTGAATTC
CCAGTTCGAATGAGCCTAGGTCGACTTAAG

Use Table 8.1 (page 203) to identify the recognition site for each of the following restriction enzymes, and hence show the cuts that would be made and state whether they produce blunt ends or sticky ends.
  a BamHI
  b EcoRI
  c HindIII
  d PvuII
19 Compare and contrast synthetic hormones and biosynthetic hormones.
20 The most commonly used test to see whether thyroid function is adequate is a blood test for thyroid-stimulating hormone (TSH).
  a How would a blood test for TSH show whether the thyroid is functioning normally?
  b A test for TSH in the blood can also be used to determine whether a person's diet has sufficient iodine. How would such a test be able to show whether iodine levels are adequate?

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21 Explain how gene therapy is different from cell therapy.

22 Imagine that you are a doctor. One of your patients is overweight and complains of feeling constantly hungry and thirsty. You suspect the patient may have type 2 diabetes.
   a What tests would you do to find out whether the person is suffering from type 2 diabetes?
   b If type 2 diabetes is positively diagnosed, what treatment would you recommend for the patient?

23 Goitre, enlargement of the thyroid gland, can be associated with both over-production and under-production of thyroid hormone. Explain how this is possible.

24 Graves' disease is caused by an abnormality of the immune system. The immune system produces an antibody that behaves in the same way as TSH. Explain how this would lead to hyperthyroidism.

Extend

25 When the Human Genome Project was launched in 1990 it was expected to take until 2005 for complete mapping to be achieved. However, the results of the project were published in 2001, four years ahead of schedule. Find out what enabled the project to advance much faster than originally anticipated.

26 Pregnant women need up to three times more insulin than normal. If the body is unable to produce that much insulin, a condition called gestational diabetes develops. Find out how gestational diabetes could affect the developing foetus.

27 The use of blood products sourced from living donors and human growth hormone from cadavers resulted in products that were devised to improve quality of life, but which also had life-threatening side effects. Using the Internet, find out the types of diseases that were involved with these contaminated products and how they affected the recipients of those products. How has recombinant DNA technology overcome these life-threatening side effects?

28 Uncontrolled diabetes may result in unconsciousness or diabetic coma. Conduct research to find out:
   a the three different types of diabetic coma and the cause of each
   b the relationship between each type of coma and the two types of diabetes
   c the first aid and treatment for diabetic coma.

29 One researcher in the United States stated:

> Tissue engineering holds out promise of truly healing the heart after congestive heart failure.… Through tissue engineering we could actually restore the function of the heart by replacing large portions of the damaged heart muscle by a bioartificial one.

This same researcher has been working for a long time on developing the ideal scaffolding to support the injected cells and the architecture of the heart. Use an Internet search engine to find out the type of scaffolding material that is being used in such research and the success that has been achieved to date.

30 The impact of biotechnology on our daily lives is growing. Much is being said and written about developments in the use of stem cells to aid the treatment of disease. Hold a class debate to canvas both sides of the question: 'Should the Australian federal government support stem cell research?' Remember to keep an open mind and to respect the opinions of others.

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31 Population projections by the Australian Bureau of Statistics indicate that by the year 2051 the proportion of the total Australian population that is aged 65 years or more will almost double. Discuss how the impact of this shift in the age structure of the population will affect diseases of ageing such as Parkinson's and Alzheimer's, with particular reference to the stress it will create for health systems and resources.

32 Recombinant DNA technology has resulted in the manufacture of far more human growth hormone than was available in the past. It is now being used to overcome some of the cosmetic effects of ageing. Find out:

- **a** when human growth hormone first became available for use with adults
- **b** what evidence there is of beneficial results from anti-ageing use of this hormone
- **c** if there are risks involved in the use of the hormone in this way
- **d** if there are other benefits to adults in the use of this hormone.

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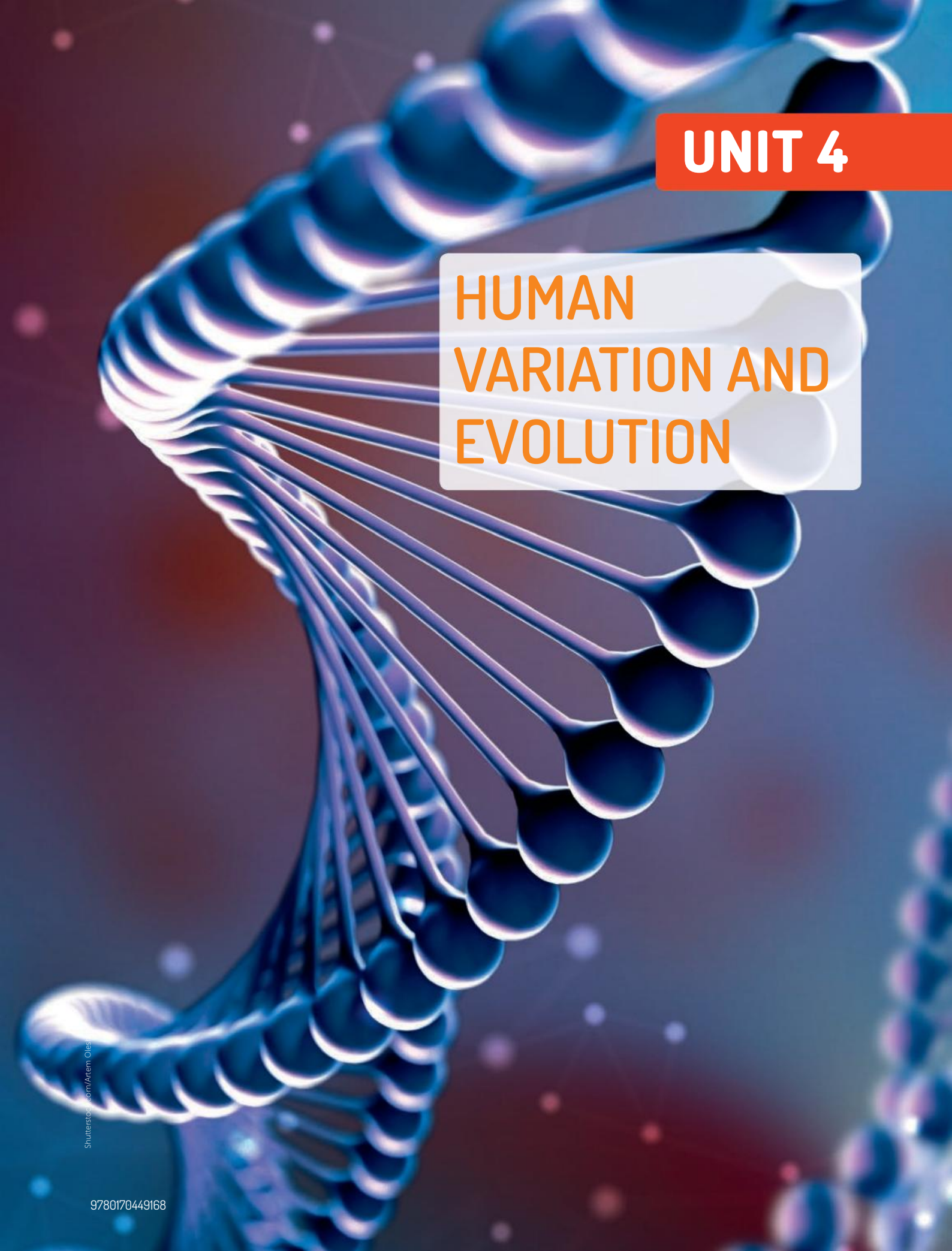

UNIT 4

HUMAN VARIATION AND EVOLUTION

9780170449168

9 EVOLUTION PRODUCES CHANGES ACROSS GENERATIONS

UNIT 4 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including the use of virtual or real biotechnological techniques of polymerase chain reaction (PCR), gel electrophoresis for deoxyribonucleic acid (DNA) sequencing, and techniques for relative and absolute dating, safely, competently and methodically for valid and reliable collection of data
- interpret a range of scientific and media texts, and evaluate models, processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments
- select, use and/or construct appropriate representations, including phylogenetic trees, to communicate conceptual understanding, solve problems and make predictions

SCIENCE UNDERSTANDING

Mutations

- mutations in genes and chromosomes can result from errors in DNA replication, cell division or from damage caused by mutagens
- different genotypes produce a variety of phenotypes, which are acted on differently by factors in the environment, producing different rates of survival
- mutations are the ultimate source of variation introducing new alleles into a population: new alleles may be favourable or unfavourable to survival

Gene pools

- populations can be represented as gene pools that reflect the frequency of alleles of a particular gene; gene pools can be used to compare populations at different times or locations
- gene pools are dynamic, with changes in allele frequency caused by:
  - mutations
  - differing selection pressures
  - random genetic drift, including the founder effect
  - changes in gene flow between adjoining groups
- the incidence of genetic diseases in particular populations illustrates the effects of different factors on the dynamics of gene pools, including the incidence of Tay-Sachs disease, thalassemia (α and β) and sickle-cell anaemia
- natural selection occurs when factors in the environment confer a selective advantage on specific phenotypes to enhance survival and reproduction
- the mechanisms underpinning the theory of evolution by natural selection include inherited variation, struggle for existence, isolation and differential selection, producing changes to gene pools to such an extent that speciation occurs

Source: School Curriculum and Standards Authority, Government of Western Australia

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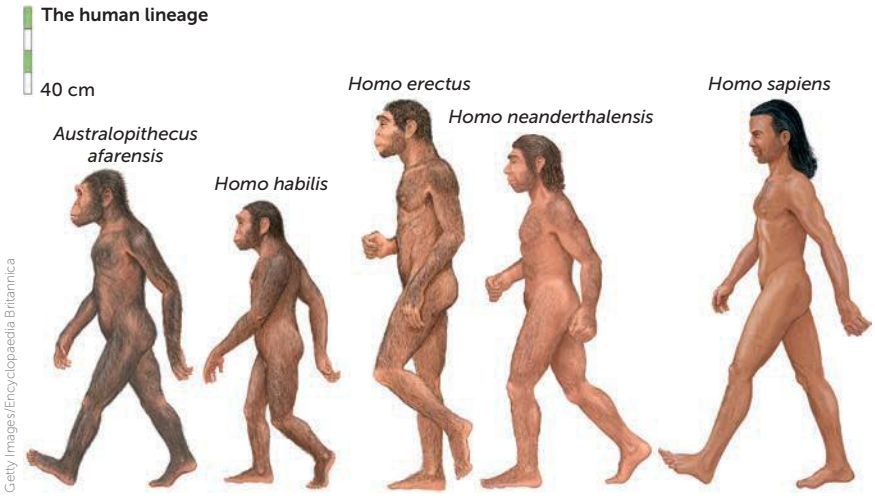

Getty Images/Encyclopaedia Britannica

FIGURE 9.1 An artist's representation of different hominin species as they have changed through evolution

In Chapter 7 you learnt about bacteria that have become resistant to antibiotics. Some bacteria may have a mutated gene that allows them to survive antibiotic treatment. These bacteria are the ones to reproduce and, therefore, there are more resistant bacteria in the next generation. Over time, more and more bacteria are resistant and so the species has **evolved**.

Evolution is the change in characteristics of a species over time. It is a gradual change that occurs over a number of generations, rather than the change of a particular individual or generation. The **phenotypes**, or set of characteristics, of individuals are a result of the alleles, or **genotype**, for each trait. Therefore, evolution reflects the changes in allele frequency in populations.

Changes to the alleles present in a population may be due to new alleles forming as a result of mutations or being introduced to a population through migration. The frequency of these alleles may alter because of a selective pressure in natural selection or by chance in genetic drift. We will be looking at these mechanisms of evolution in this chapter.

Antibiotic resistance
This article has more information about bacteria evolving to become antibiotic resistant through natural selection.

9.1 MUTATIONS

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FIGURE 9.2 Variation occurs in humans

Gene pool

A **population** is a group of organisms of the same species living together in a particular place at a particular time. When studying populations, **geneticists** – scientists who specialise in the study of inheritance – prefer to consider the characteristics of the population as a whole and not those of the individuals that make up the population. They find it convenient to pool the genotypes of all the individuals capable of reproducing and refer to this as the gene pool. Thus, the **gene pool** is the sum of all the alleles in a given population.

When studying a population, geneticists are interested in how often each allele of a gene occurs in the gene pool for that population. These are called the **allele frequencies** for the population. For example, an allele for cystic fibrosis is found on chromosome number 7. If the frequency of the cystic fibrosis allele in a given population is 5%, then among population members, five in every 100 of chromosome 7 will carry that allele. Ninety-five out of 100 chromosome 7s will have the normal form of the gene.

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FIGURE 9.3 The frequency of the allele for blue eye colour would be higher in the Scandinavian population than in the African population

Populations that differ in the characteristics they possess are likely to have different frequencies of the various alleles of a gene in their respective gene pools. For example, Scandinavians commonly have blue eyes, whereas black Africans have brown eyes. The frequency of the allele for blue eyes would be much higher in the Scandinavian gene pool than in the African gene pool. Thus, any two populations having differing characteristics are likely to have different gene pools.

Mutations

Offspring may show variations that do not resemble either parent, and have never occurred before in the history of the family. Therefore, they are due not to an allele being passed down from the parents, but to a new allele being formed. This can happen when the DNA is changed by a **mutation**, resulting in a different variation of the trait. Not all mutations are harmful, but many are. An organism with a characteristic resulting from a mutation is called a **mutant**. There are two main types of mutations:

- **gene mutations**, which are changes in a single gene so that the traits normally produced by that gene are changed or destroyed
- **chromosomal mutations**, in which all or part of a chromosome is affected.

If a mistake occurs spontaneously when the DNA molecule is copied during mitosis or meiosis, or when the chromosomes are separated during meiosis, the change may have significant effects on the functioning of the cell. However, many mutations are repaired, and therefore don't cause a problem. If they do remain, when the cell divides the mutated DNA will be copied and passed on to daughter cells. If the daughter cells are gametes, the mutation may be passed on from generation to generation.

There are relatively few mutations in human populations, considering the millions of cell divisions that occur. Those that do occur sometimes result in traits better suited to a particular environment, and so may contribute to human survival.

Mutations
This website provides more information on mutations.

Causes of mutations

Mutations occur without any known cause, but a number of agents are known to increase the rate at which they occur. These are called **mutagenic agents** or **mutagens**. Some known mutagens are mustard gas, formaldehyde, sulfur dioxide and some antibiotics. All kinds of ionising radiation, including ultraviolet light, X-rays, cosmic rays, radiation from radioactive waste, and fallout from atomic and nuclear explosions, are also mutagenic. If a woman is treated with large doses of X-rays during the first three months of her pregnancy, the child may be born with intellectual disability, skeletal malformations, or microcephaly (a condition where the head is small in relation to the rest of its body). For this reason, doctors try to avoid using X-rays early in pregnancy.

Activity 9.1
Investigating the effect of ultraviolet radiation on UV-sensitive and wild-type forms of the yeast *Saccharomyces cerevisiae*

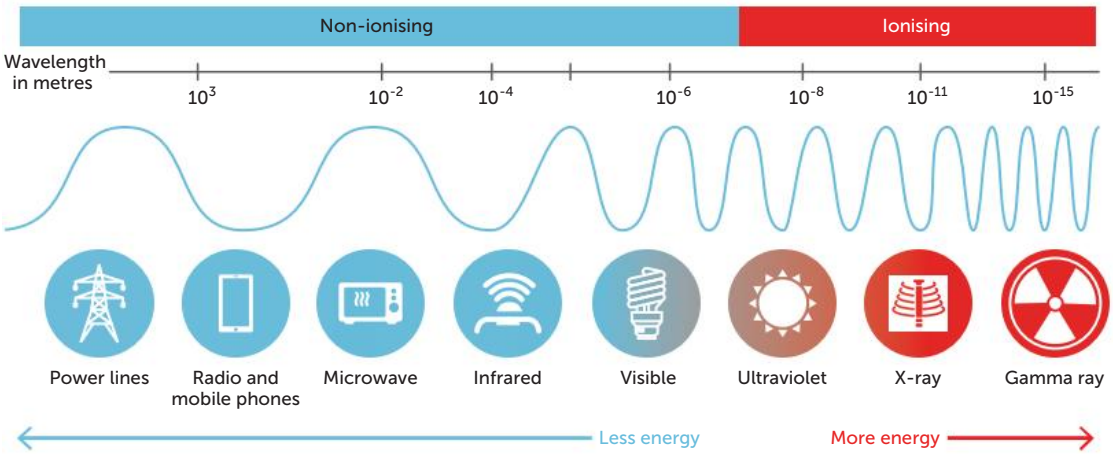

FIGURE 9.4 Ionising radiation is an example of a mutagen

Key concept

Mutations are changes in the DNA resulting in a variation in the associated trait. They may occur spontaneously, but are often due to exposure to a mutagen.

Types of mutations

DNA is composed of a double helix, each side of which is a long string of four types of nucleotides. Each nucleotide possesses identical sugar–phosphate groups that contribute to the DNA framework but differs in the base that links the two frameworks. Within genes, the sequence of the bases in the DNA is the code for the amino acids used to build a protein. Each group of three bases codes for an amino acid.

When it was recognised that genetic information is contained in the sequence of bases in the DNA, it became possible to understand the chemical nature of gene mutations. A change in the bases could change the amino acid and so could alter a protein. It is possible that a mutation could have no effect at all, or it may alter the protein or prevent it from being produced. Thus, if the DNA of a particular gene is altered, the protein for which it codes may be missing or abnormal. Just one missing or abnormal protein can have an enormous effect on the entire body.

Albinism, for instance, is the result of one missing protein. **Albinism** is marked by an absence of pigment from the hair, skin and eyes. The hair of a person with albinism tends to be whitish blond, the skin extremely pale and the eyes pinkish.

iStock.com/Vesnaandjic

FIGURE 9.5 Albinism is an inherited condition caused by a mutation that results in just one missing protein

Mutations can be classified by a number of different characteristics. It is the sum of these characteristics that determines the overall impact the mutation will have on the individual.

Cause of the mutation

You have already learnt that mutagens in the environment can increase the chance of mutations occurring. These mutations are known as **induced mutations**. Other mutations occur due to a random error in a biological process such as mitosis or meiosis. These are called **spontaneous mutations**.

Heritability of the mutation

One way of classifying mutations is by the type of cells where they occur, such as a person's body cells or reproductive cells. When the body cells, or somatic cells, are involved with a mutation, it is known as a **somatic mutation**. In this situation, only the individual with the somatic mutation is affected. Each time the mutant body cell divides, the mutation is passed on to the daughter cells. However, as the reproductive cells are not affected, once the individual dies the mutation is lost. Somatic mutations are involved in many cancerous growths that may be a result of a mutagenic agent.

If the reproductive cells are affected, the mutation can occur in the gametes and may then be passed on to the next, and subsequent, generations. These are known as **germinal** or **germline mutations**. In this case, the individual in whom the mutation occurs is not usually affected. However, that individual produces gametes with changed DNA. If conception occurs involving one of the affected gametes, the embryo is often naturally aborted early in the pregnancy. Diseases such as **phenylketonuria (PKU)** can arise through a mutation during the formation of gametes and can be passed on to offspring.

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FIGURE 9.6 Lead aprons are used to protect the cells, particularly gametes, during exposure to ionising radiation

Effect of the mutation

Another way that mutations can be classified is based on their effect.

- **Missense mutations** cause a change in the amino acid, and therefore in the protein produced.
- **Nonsense mutations** change the base sequence to the code to STOP. This means that the synthesis of the protein will stop, and so a shorter protein is produced that is unlikely to be able to fulfil its function.
- **Neutral mutations** cause a change in an amino acid; however, the amino acid is of the same type and does not change the structure of the protein enough to change its function.
- **Silent mutations** do not cause any change in the amino acid, and therefore in the protein produced. This is possible, as most amino acids are coded for by more than one base sequence.

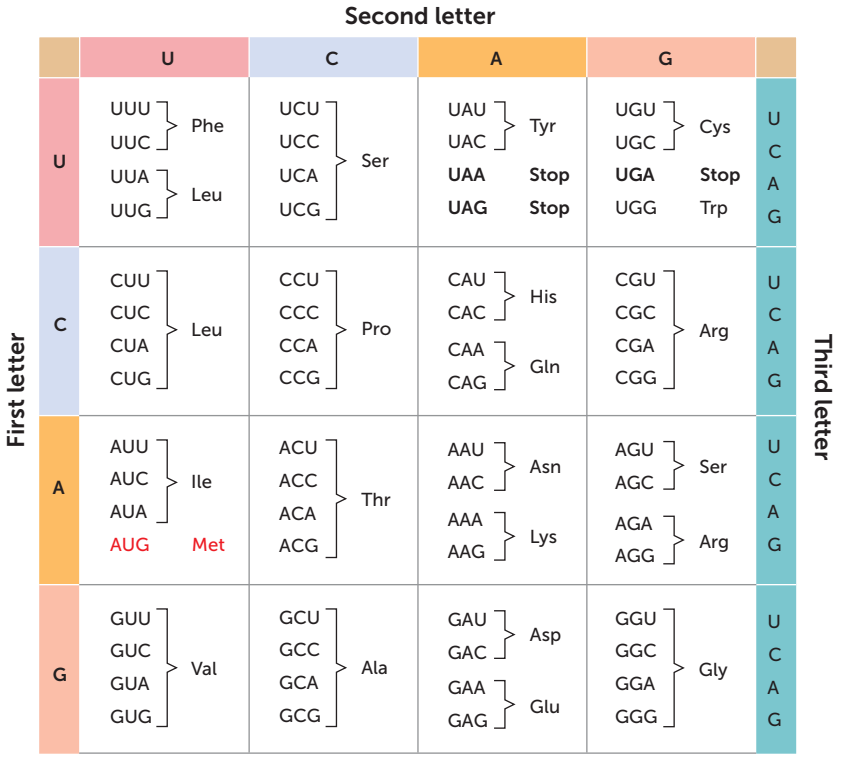

FIGURE 9.7 The codons that code for each amino acid. Note that the codon is the sequence of three bases on mRNA. This will be complementary to the base sequence on the DNA molecule

Extent of the mutation

Another characteristic that is used to classify mutations is the amount of DNA affected. This could range from a single base to a whole chromosome. A gene mutation affects only a single gene, while a chromosomal mutation affects a number of genes. It may change the structure of a chromosome or the number of chromosomes. Chromosomal mutations often cause abnormalities so severe that miscarriage often occurs early in the pregnancy.

Change in the DNA

Mutations vary in the change in the DNA. **Point mutations** are due to changes in a single nucleotide; therefore, only one base is changed. These mutations may be due to a nucleotide being:

- *inserted* – a new nucleotide is added to the DNA strand
- *substituted* – an existing nucleotide is replaced with another one, with a different base
- *deleted* – a nucleotide is removed from the DNA strand.

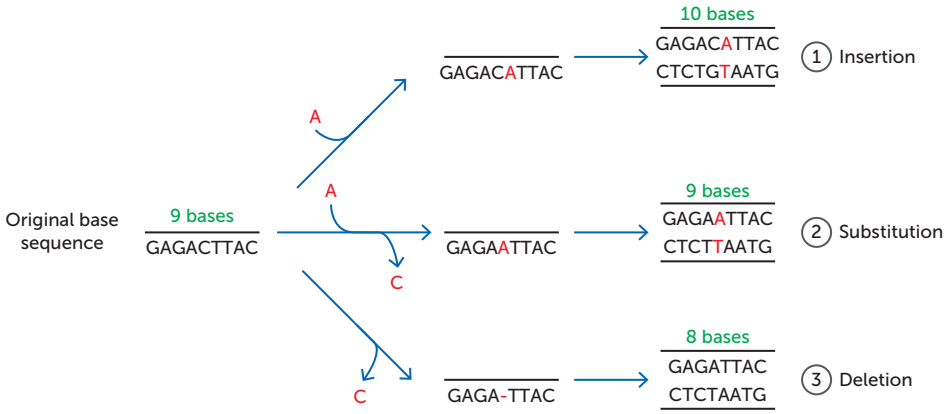

FIGURE 9.8 Insertion, substitution and deletion mutations

Some mutations will result in a frameshift. A **frameshift** occurs when bases have been added or removed. This results in the series of three bases that code for an amino acid starting at a different base. Therefore, although the mutation may have only altered a single base, frameshift mutations affect the outcome for all the DNA from that point on.

Frameshift mutations will not occur when three bases are added or deleted. In these instances, the DNA will simply code for one more, or one less, amino acid, but the rest of the amino acids will be the same. Therefore, it would still be a mutation, just not a frameshift mutation.

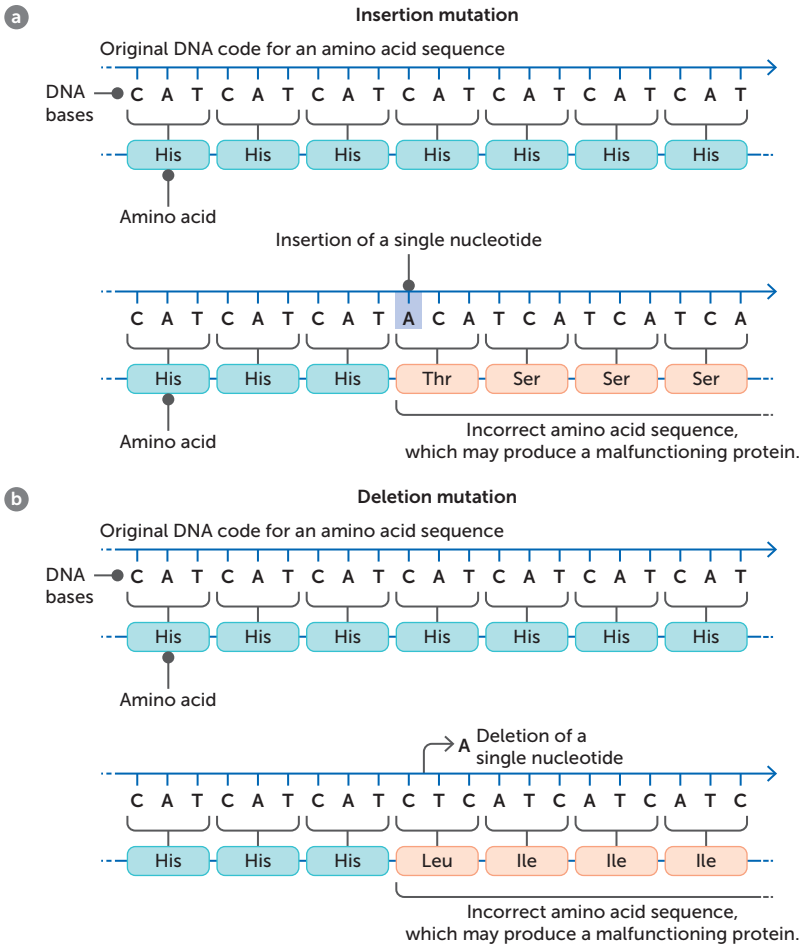

FIGURE 9.9 Frameshift mutations due to the **a** insertion and **b** deletion of a nucleotide

Other mutations affect a larger section of DNA. They may be due to:

- *duplication (or insertion)* – a section of chromosome occurs twice
- *deletion* – a piece of DNA is removed
- *inversion* – breaks occur in a chromosome and the broken piece joins back in, but the wrong way around
- *translocation* – part of a chromosome breaks off and is rejoined to the wrong chromosome
- *non-disjunction* – during meiosis, a chromosome pair does not separate and so one daughter cell has an extra chromosome and one daughter cell has one less than the normal number. These are sometimes referred to not as 'mutations', but as **aneuploidy** – a change in the chromosome number.

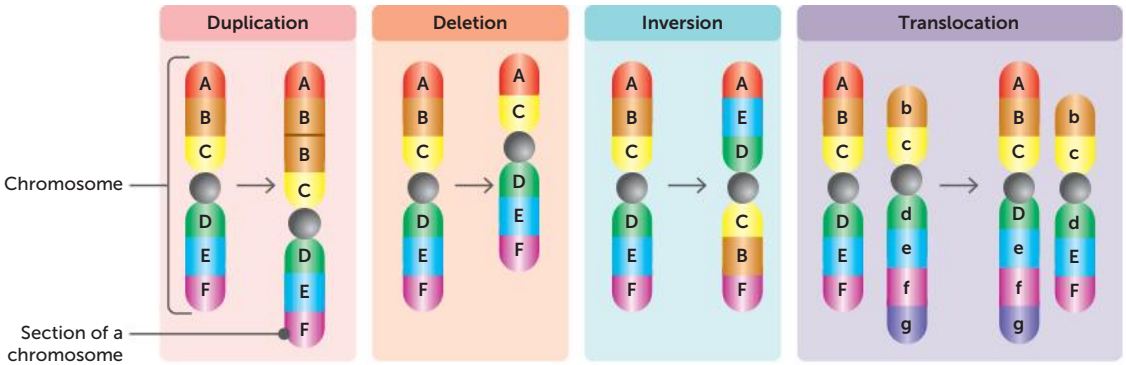

FIGURE 9.10 Mutations due to duplication, deletion, inversion and translocation of sections of DNA

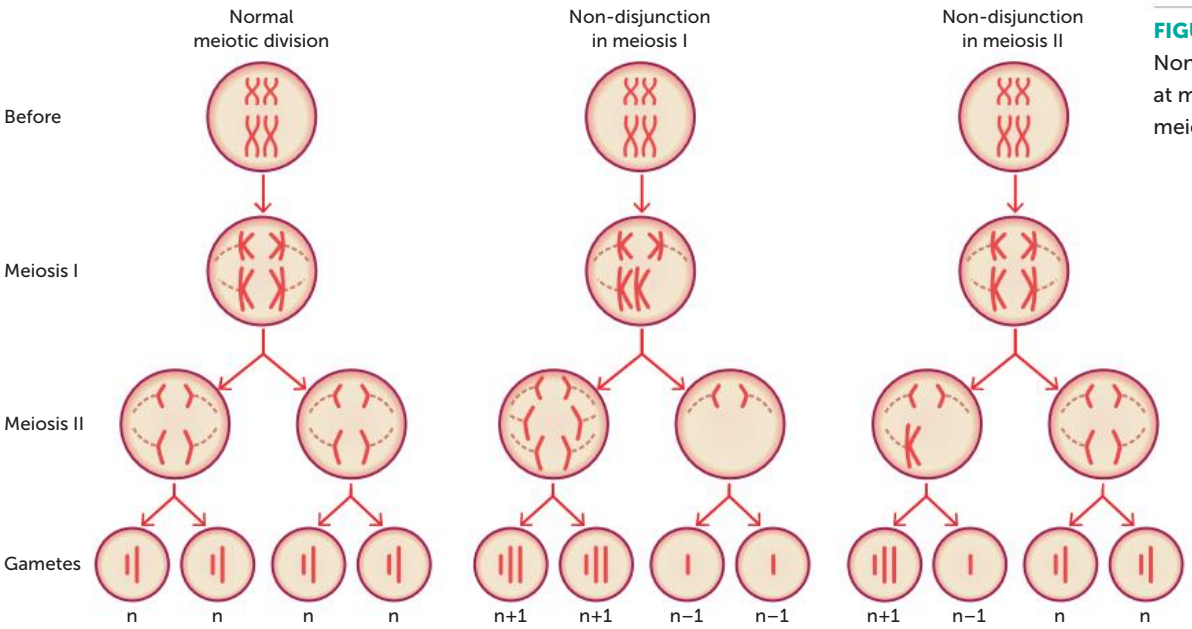

FIGURE 9.11 Non-disjunction at meiosis I and meiosis II

Key concept

Mutations can be classified based on their cause, where they occur, their effect, the amount of DNA they affect and the change in the DNA.

Conditions due to mutations

Gene mutations

Duchenne muscular dystrophy may occur through gene mutation. This may arise through a mutation in the mother, which can then be inherited by her sons. The mutation may also occur in a male zygote so that the child develops the disease. This disease results in a wasting of the leg muscles and later the arms, shoulders and chest. Duchenne muscular dystrophy usually becomes apparent around the age of three to five years, when muscle weakness becomes evident. Eventually, death occurs due to failure of the respiratory muscles. Boys with the Duchenne form of muscular dystrophy are unlikely to live for more than 20–25 years.

Cystic fibrosis is another genetically determined disease caused by a mutation. The mutation occurs in a huge gene on chromosome number 7. The gene has the code for 1480 amino acids that make up a protein that regulates the passage of chloride ions across the cell membrane. Without the correct protein the affected person suffers from a variety of symptoms: salty-tasting skin; persistent coughing, wheezing or pneumonia; and digestive and other problems. The mutant allele is recessive, so a sufferer must inherit it from both parents.

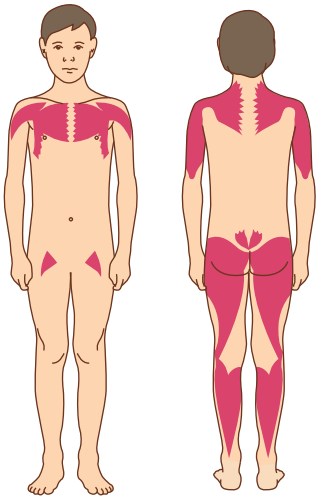

FIGURE 9.12 The muscles that are affected by Duchenne muscular dystrophy

Chromosomal mutations: Trisomy

Trisomy is a result of non-disjunction, failure of one or more chromatids to separate in the second division of meiosis. The eggs or sperm formed when non-disjunction occurs have one chromosome too many, or one chromosome is missing.

A chromosomal mutation that occurs relatively frequently, especially in children of older mothers, is **Down syndrome**, or **trisomy 21**, where the child has three of chromosome 21 instead of the normal two. People with Down syndrome have a characteristic facial expression, intellectual disability and weak muscles. They may also suffer from some birth defects such as heart defects or digestive abnormalities.

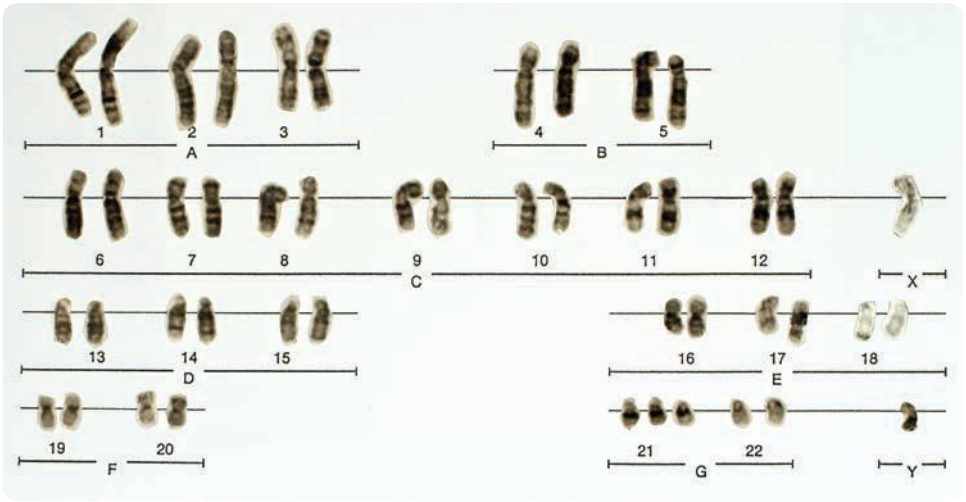

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FIGURE 9.13 Karyotype of Down syndrome (note the extra chromosome 21)

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FIGURE 9.14 Down syndrome children have a characteristic facial expression

Many of the symptoms of Down syndrome can also occur when part of an extra copy of chromosome 21 is attached to one of the other chromosomes. This is called partial trisomy.

Trisomy also occurs with other human chromosomes. **Patau syndrome** is when an extra chromosome 13 produces individuals with intellectual disability, microcephaly, an extra finger on each hand, a cleft palate and/or cleft lip, and malformations of the ears and eyes.

The extra chromosome 13 can come from either the mother's egg cell or the father's sperm cell. The features of trisomy 13 result from having this extra chromosome in each of the body's cells. Trisomy 13 occurs in about one out of every 5000 live births. However, more than 80% of children with trisomy 13 die within a month of birth.

Trisomy can also occur with the sex chromosomes. In males, non-disjunction may occur during either the first or the second meiotic division, producing individuals with either an extra X chromosome (*XXY*) or an extra Y chromosome (*XYY*). Individuals with trisomy *XXY* are normal as boys but develop **Klinefelter syndrome** as adults. They have small testes that do not produce sperm, the breasts are enlarged and body hair is sparse. Occasionally, the individual has an intellectual disability.

Chromosomal mutations: Monosomy

Monosomy is where an individual is missing a chromosome. If an autosome is completely missing, monosomy usually results in severe malformations and miscarriage. If only part of a chromosome is missing, it is referred to as **partial monosomy**. Part of the chromosome has two copies, but part has only one copy.

An example of partial monosomy is **Cri-du-chat syndrome** (from the French for 'cry of the cat'), a rare genetic disorder due to a missing portion of chromosome 5. The syndrome gets its name from the characteristic cry of infants born with the disorder. The infant sounds just like a meowing kitten, due to problems with the larynx and nervous system.

Monosomy can also occur with the sex chromosomes. Individuals with a chromosome set with only one *X* chromosome (monosomy *X*) suffer from **Turner syndrome**. These females are short in stature, lack secondary sexual characteristics and are infertile.

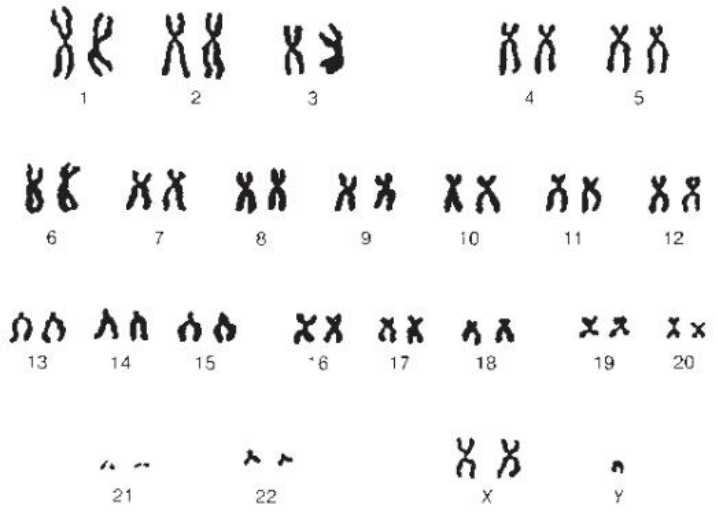

FIGURE 9.15 Karyotype for Klinefelter's syndrome

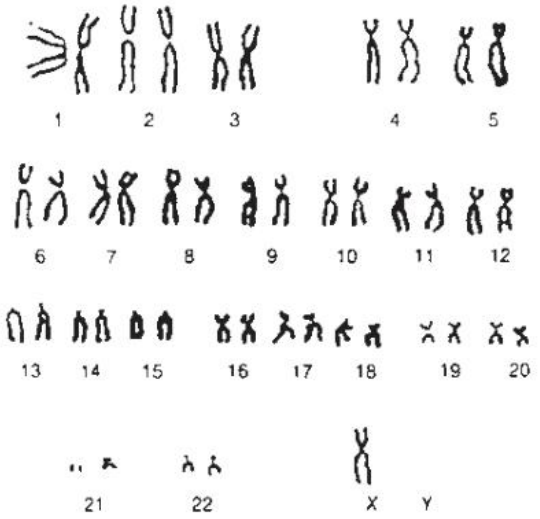

FIGURE 9.16 Karyotype for Turner syndrome

Lethal recessives

Most gene mutations produce a recessive allele because they prevent the gene from producing a protein that will be able to function in the body. A person could therefore have large numbers of mutations in the genes and be totally unaware of them. If the person reproduces with a partner who has the same recessive mutation, the recessive condition could appear in their offspring. This is what happens when couples unexpectedly have a child with cystic fibrosis.

Some recessive mutations are lethal if they are not masked by a dominant normal allele. These **lethal recessives** cause the death of the embryo or foetus by a miscarriage or spontaneous abortion, or the early death of the child.

It is easy to see how a lethal recessive mutation could cause changes in the composition of a gene pool. People who inherit two such alleles would die before their alleles could be passed on to the next generation, so the proportion of lethal recessive alleles in the gene pool would gradually be reduced.

Tay-Sachs disease (TSD) is a disorder of lipid metabolism that is inherited in an autosomal recessive pattern. It is a lethal recessive condition caused by a mutation in the HEXA gene that codes for the enzyme beta-hexosaminidase. This enzyme is responsible for breaking down toxic substances, including a fatty substance called GM2 ganglioside, in the brain and spinal cord. The missing enzyme results in the accumulation of GM2 ganglioside in the nervous system, which destroys the neurons. A baby with two recessive alleles for TSD develops normally for the first few months, and then deterioration that causes intellectual and physical disabilities begins. Death usually occurs in early childhood.

Examples of unbalanced chromosomal arrangements
This website provides more information on some chromosomal mutations.

WS
9.1 Mutations

Questions 9.1

RECALL KNOWLEDGE

1. Define 'mutagen' and list three examples.
2. Describe the effects of these different types of mutation:
   - **a** missense mutation
   - **b** nonsense mutation
   - **c** neutral mutation
   - **d** silent mutation.
3. Draw and label diagrams to demonstrate how an inversion changes the base sequence on DNA.
4. Explain why only germline mutations are passed on to the next generation.
5. List two conditions due to:
   - **a** gene mutations
   - **b** chromosomal mutations.

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6 Is Down syndrome an example of a gene mutation or a chromosomal mutation? Explain your answer.

APPLY KNOWLEDGE

7 Use a diagram to explain why the insertion of a single nucleotide can cause a frameshift mutation, resulting in a change in many amino acids.

8 The original base sequence of a small section of DNA is shown below.

C	C	T	A	G	T	C
G	G	A	T	C	A	G

Name the point mutation that has occurred in each of the following.

a

C	C	A	A	G	T	C
G	G	T	T	C	A	G

b

C	C	T	A	G	G	T	C
G	G	A	T	C	C	A	G

c

C	T	A	G	T	C
G	A	T	C	A	G

9 Use Tay-Sachs disease as an example to explain how a gene mutation can be lethal.

10 Some mutations result in a STOP codon, preventing any more amino acids being added to the chain. These are known as nonsense mutations. Figure 9.7 shows the base sequence on the mRNA, which is a copy of the coding strand of the DNA but containing the base uracil instead of thymine. Describe the mutation that could have occurred to each of the following original base sequences on the coding strand to result in a nonsense mutation.

a AAA
b TCGA
c TAT

9.2 MIGRATION

Changes in allele frequencies in a gene pool can also be due to gene flow brought about by migration. **Gene flow** is the movement of genetic material from one population to another. When individuals move between populations, they enable gene flow. This movement is known as **migration**. Therefore, if immigrants to a certain country bring alleles that are not already in the population, the frequencies for the alleles of that gene will be altered. This has occurred in China, for example. In the past, the Chinese population all had the Rh-positive blood group. The Rh, or Rhesus factor, is an antigen found on the surface of red blood cells. People with this antigen are referred to as Rh+; those who do not have the antigen are Rh−. When European countries began trading with China in the 16th century, European immigrants and sailors introduced the Rh− allele into the Chinese population. However, the frequency of the allele is still very low in China compared to other countries.

An example of how the distribution of ABO blood groups has been influenced by migration is the change in the frequency of the I^B allele across Europe and Asia. The inhabitants of East Asia, the Mongols, have a proportionately higher frequency of the allele I^B than those living to their west in Europe. In fact, it is thought that most Western Europeans originally did not have the I^B allele at all. In the 12th and 13th centuries, the Mongols invaded Europe on a number of occasions, spreading not only their culture but their genes as well. Today, there is a steady decrease in the I^B allele from Central Asia to Western Europe. Interestingly, the lowest concentrations of the I^B allele are now in the Pyrenees mountains and a few isolated locations in Scandinavia.

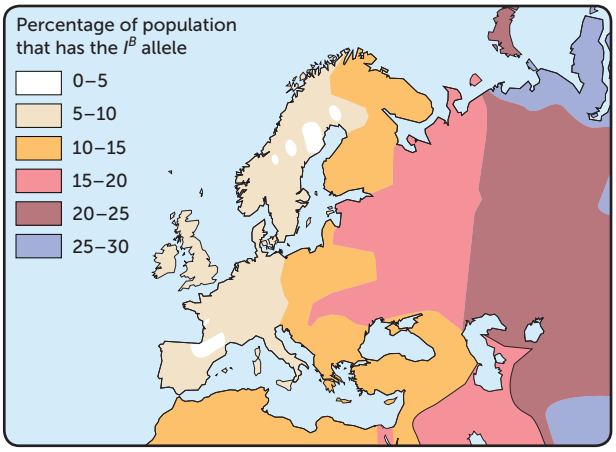


FIGURE 9.17 Distribution of the allele for the B blood group in Europe

Barriers to gene flow

Populations are often kept apart by barriers that inhibit the amount of interbreeding between them. This isolation leads to separate gene pools forming. Barriers to gene flow can be classified based on their cause.

- **Geographical barriers** include oceans, mountain ranges, large lake systems, deserts and expansive ice sheets. For example, the original inhabitants of Australia were isolated for thousands of years by ocean barriers that formed as sea levels rose.
- **Sociocultural barriers** such as economic status, educational background and social position are barriers to interbreeding. For example, statistics indicate that Australians tend to marry people of similar educational background, and members of particular religious groups favour partners who have the same faith. Religion and language can also be barriers to gene flow. Some religions do not allow marriages outside the religion, and it is unlikely that people who cannot communicate with one another will marry.

Key concept

Gene flow is the movement of genetic material from one population to another. It is facilitated by migration, but hindered by barriers such as geography and sociocultural factors.

Questions 9.2

RECALL KNOWLEDGE

1. Define 'gene flow'.
2. Describe how migration facilitates gene flow.
3. List four barriers to gene flow.
4. Describe how religion may be a barrier to gene flow.

APPLY KNOWLEDGE

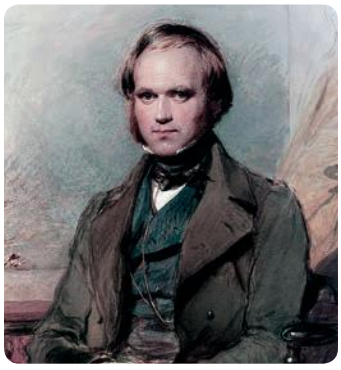
5. Explain why geographical barriers have less influence on gene flow in today's populations than in previous times.
6. Describe an example where barriers to gene flow would be:
   - a an advantage
   - b a disadvantage.

9.3 NATURAL SELECTION

Development of the theory of evolution

There are countless millions of species of plants, animals and micro-organisms living on Earth today. How has this multitude of species come into existence? Until the 1800s, it was widely believed that God, or a supreme being, had individually created each species. This is known as **special creation** and it is still the belief of members of some religious groups. Evolution is a gradual change in the characteristics of a species. The theory of evolution through natural selection was put forward independently by Charles Darwin and Alfred Russel Wallace in 1858. However, it is Darwin's name that is usually associated with this theory because of the massive amount of supporting evidence he collected.

Darwin was a keen amateur naturalist and as a young man he joined a surveying expedition as its biologist. He voyaged on HMS *Beagle*, visiting, among other places, the Galapagos Islands, New Zealand and Australia. This voyage, and the material Darwin



Alamy Stock Photo/FineArt

FIGURE 9.18 Charles Darwin. Together with Alfred Russel Wallace, Darwin put forward the theory of evolution through natural selection in 1858

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collected, was to be the preparation for all his later work. The Galapagos Islands were especially important for his research. On these islands, Darwin was able to observe the differences and similarities between animals separated by:

- geography – those living on the mainland of South America and those on the various islands
- time – animals recently extinct and species still alive.

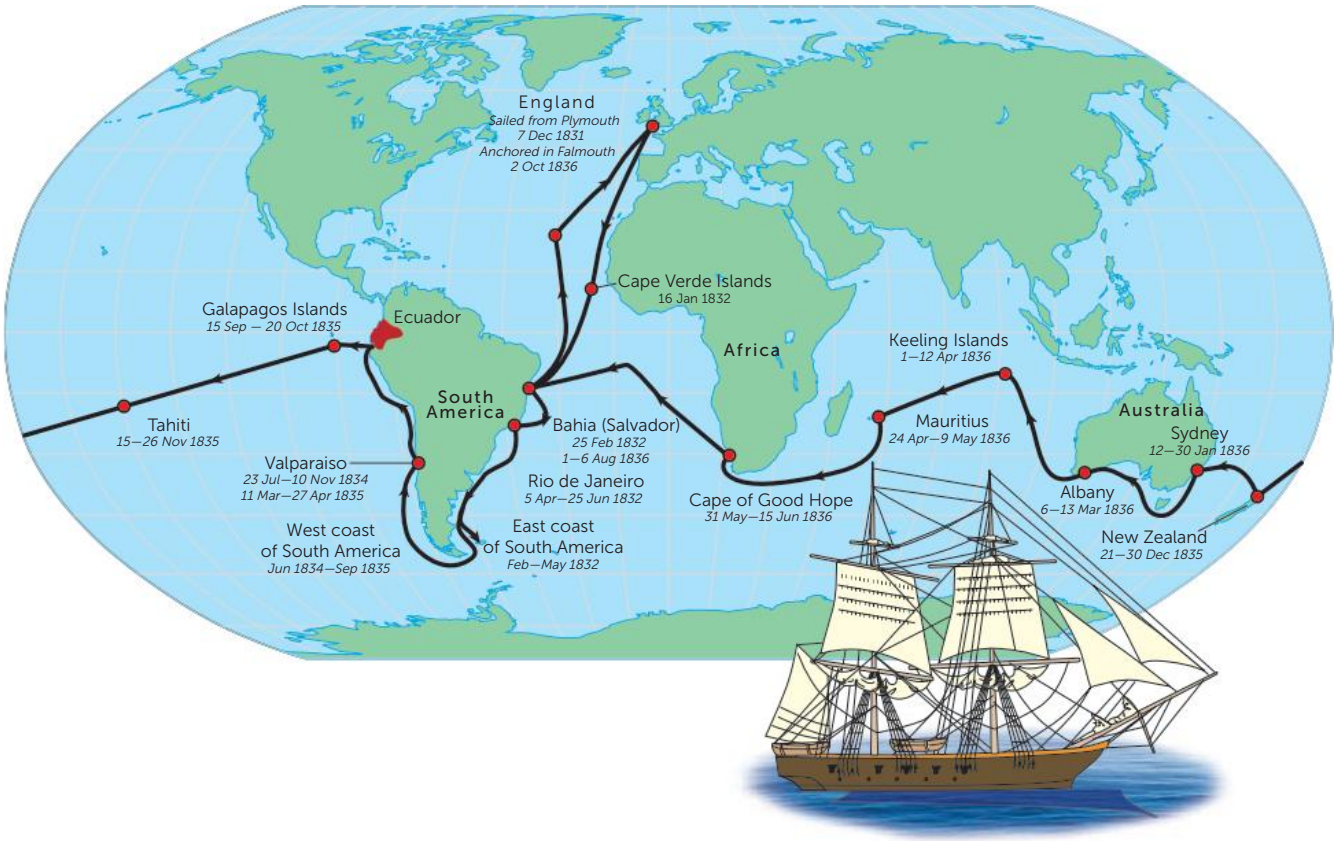

FIGURE 9.19 The route taken by HMS *Beagle* from December 1831 to October 1836. Darwin's observations on this journey were crucial to his later ideas on natural selection

These observations led Darwin to question the commonly held belief that living species had always been exactly the same as they then appeared. He became convinced that species did change. But how did the changes take place?

Darwin was greatly influenced by the works of other people. Carolus Linnaeus (1707–78) established the basis of our present system of classification and the binomial system of naming organisms using the generic (genus) and specific (species) names. This system was important to Darwin as it enabled him to classify and organise the material he collected.

Another major influence on Darwin was a book, *The Principles of Geology*, written by his friend Charles Lyell. Lyell hypothesised that the natural forces existing in the past were much the same as those existing in his own time. This hypothesis implied that Earth's surface had been gradually moulded over a very long period of time, by such simple forces as changes in temperature, running water and earth movements. Lyell's ideas provided Darwin with a concept of constant change against which he could view his own work.

Thomas Malthus, a British clergyman and political economist, provided the idea for the foundation of Darwin's theory of natural selection. Malthus, in 'An Essay on the Principle of Population', pointed out that the human population was increasing at a rate far exceeding the rate of food production. Drawing on examples from natural populations of plants and animals, he demonstrated that natural reproduction rates exceeded the available resources; that is, more plants

and animals are produced than can possibly survive. Darwin realised that under these circumstances a struggle for existence would occur, with favourable variations being preserved and unfavourable ones being gradually lost from the population.

In 1858, Darwin received a copy of an essay by Alfred Russel Wallace, a naturalist then on the island of Ternate in Indonesia (then the Dutch East Indies). Wallace's essay, 'On the Tendency of Varieties to Depart Indefinitely from the Original Type', covered the same ideas that Darwin had been working on. Darwin had been collecting evidence and refining his ideas for 20 years, but Wallace's essay was the stimulus for him to publish his views. A joint essay was prepared by Darwin and Wallace and read before the Linnean Society in 1858.

A year later, Darwin published his first book, *On the Origin of Species*. The book created a storm of controversy, but with the support of other scientists Darwin's ideas became firmly established.

Darwin's theory of natural selection

Darwin's theory of **natural selection** was based on three observations.

- *Variation:* Darwin noted that all members of a species vary. He made no attempt to explain the source of this variation. However, he did point out that these variations were passed on from one generation to the next, with characteristics displayed by the parents being passed on to their offspring.
- *Birth rate:* Inspired by Malthus, Darwin realised that all living organisms reproduce at a rate far greater than that at which their food supply and other resources increase. This would normally result in overcrowding.
- *Nature's balance:* Darwin observed that, although the birth rate of organisms was very high, each species' numbers tended to remain at a relatively constant level.

From these three observations, Darwin made a number of interpretations. First, he realised that, because of the excessive birth rate and limited resources, there must be a **struggle for existence**; and, second, because there was a range of variations in any species, those with characteristics best suited to their environment were more likely to survive. This second point became known as **survival of the fittest**: organisms with favourable characteristics survived, while many of those with unfavourable characteristics died before they had an opportunity to reproduce and pass on the trait.

Charles Darwin
This website provides an interesting perspective on Charles Darwin and the impact of his work.

Survival of the fittest is possible because there is **variation** within any species. That is, the members of a species differ from one another in their physical characteristics, body functioning and behaviour.

With knowledge of the mechanisms of inheritance, scientists building on the work of Darwin were able to explain the process of natural selection far more satisfactorily. Today, natural selection can be viewed as the selection of those alleles in a population that give an organism a greater survival advantage. The environmental factor acting on the population is known as the **selective agent**. Those organisms that survive will pass on favourable alleles to their offspring. Gradually, over a period of time, the characteristics of a population change so that it becomes better suited to its environment. In addition, where the environment is gradually changing, characteristics that enhance survival enable succeeding generations to gradually adapt to it. An important point to note is that individual organisms do not adapt. Instead, the species adapts to its environment by natural selection, and the process of adaptation takes many generations.

Alleles and natural selection

Natural selection can be looked at in terms of the frequencies of alleles in the gene pool of a population. If the environment tends to favour a particular characteristic, more of the alleles for that trait will be passed on to the next generation. This will result in a change in the frequency of that allele in the gene pool. Over time, that characteristic becomes more frequent in the population.

Activity 9.2
Venusians: investigating natural selection

The principles of evolution through natural selection can be summarised as follows:

- There is variation of characteristics within a species.
- More offspring of a species are produced than can possibly survive to maturity.
- Because of excessive birth rate and limited resources, there is a struggle for existence or competition for survival.
- Individuals with characteristics best suited to the environment have more chance of surviving and reproducing. This is known as survival of the fittest.
- Favourable characteristics are passed on to the next generation.
- In the gene pool, the proportion of alleles that produce favourable characteristics gradually increases.

Activity 9.3
Modelling natural selection

Key concept

Natural selection is the change in allele frequency in populations as a result of a selective agent.

Examples of natural selection

Body stature

Initially, the human gene pool would have contained alleles for a whole range of statures, from the short-bodied, long-limbed physique of present-day black Africans, to the long-bodied, short-limbed stature of the Inuit people of today. Individuals with long bodies and short limbs have a smaller surface area in relation to body volume than those with short bodies and long limbs. Such individuals lose less heat in very cold environments and would therefore have a survival advantage. When individuals of this type reproduced, they would have passed on the alleles for long bodies and short limbs to their children. They, too, would have had a survival advantage and would pass on the favourable alleles to their offspring. As fewer of the short-bodied, long-limbed individuals would survive in the extreme cold, fewer of the alleles for these characteristics would have been passed on. Many individuals with less-favourable characteristics would have died before reproductive age, so the frequency of unfavourable alleles in the gene pool would gradually decrease. Over time, those alleles would have decreased to such an extent that the unfavourable characteristics would no longer occur in the population. In this way, the frequency of alleles controlling body stature in the population would have changed. Those controlling long bodies and short limbs would have increased, while those for short bodies and long limbs would have decreased. Thus, evolution or genetic change has taken place. Within a particular gene pool, the frequencies of the alleles have changed over time.

a

Alamy Stock Photo/wanderluster

b

Getty Images/Mitch Kezar

FIGURE 9.20
a Short limbs and long bodies would have evolved in very cold climates, such as where Inuits live; **b** Long limbs and short bodies would have evolved in very hot climates, such as where Africans live

Sickle-cell anaemia

The incidence of **sickle-cell anaemia** in different parts of the world is another example of natural selection operating in human populations. The *Anopheles* mosquito, which transmits the malarial parasite, is not normally an inhabitant of tropical forests. It needs quiet, stagnant pools of water for breeding sites. This habitat is more often found in open areas. As humans began to clear the forests of Africa for agriculture, they changed the environment in a manner that created additional breeding areas for *Anopheles* mosquitoes. The increased food supply from agricultural production allowed the human population to increase, providing more bodies on which the mosquitoes could feed. Thus, the incidence of malaria increased. Figure 9.21 shows the distribution of malaria throughout the world.

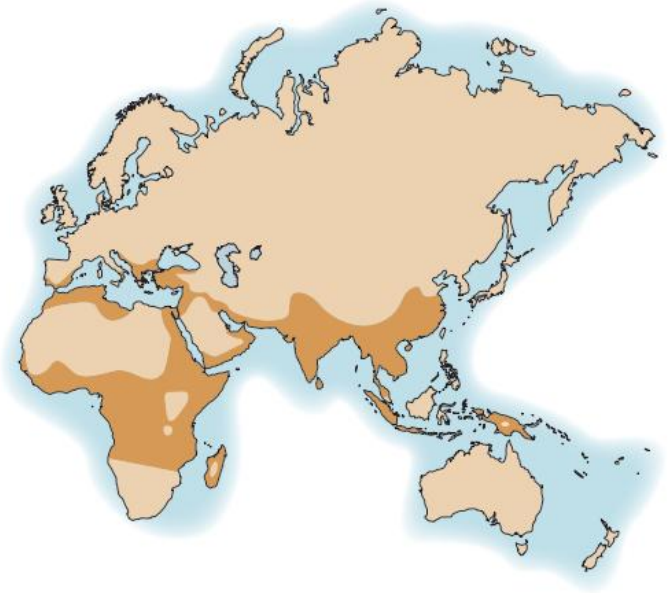

FIGURE 9.21 The distribution of malaria

In 1910, a young West Indian student living in Chicago visited his doctor with a variety of symptoms, including clogged blood vessels, pneumonia, rheumatism, heart disease, inflammation of the hands and feet, and anaemia. His doctor took a blood sample and observed it under a microscope. When air was excluded from the sample, the red blood cells showed a dramatic change in their shape from round to a crescent-like, or sickle, shape.

Subsequent investigation showed that sickle-cell disease, or sickle-cell anaemia, results when a person is homozygous for a particular recessive allele. We now know that this allele is due to a point mutation in the DNA sequence of the HBB gene. This gene codes for one of the beta-globulin proteins that make up haemoglobin in the red blood cells. The different base sequence means that the amino acid valine is added instead of glutamic acid. This results in a different form of the protein, altering the haemoglobin produced which distorts the shape of the red blood cell.

People who are heterozygotes normally show no ill effects unless oxygen is in short supply. When this occurs, their red blood cells show mild sickling. These individuals are carriers and are said to have sickle-cell trait. Individuals homozygous for the normal dominant allele have blood that shows no signs of the sickling phenomenon.

Science Photo Library/Eye of Science

FIGURE 9.22 The crescent shape of red blood cells of someone with sickle-cell anaemia

Sickle-cell anaemia
These websites provides more information on the changes to haemoglobin with sickle-cell anaemia.

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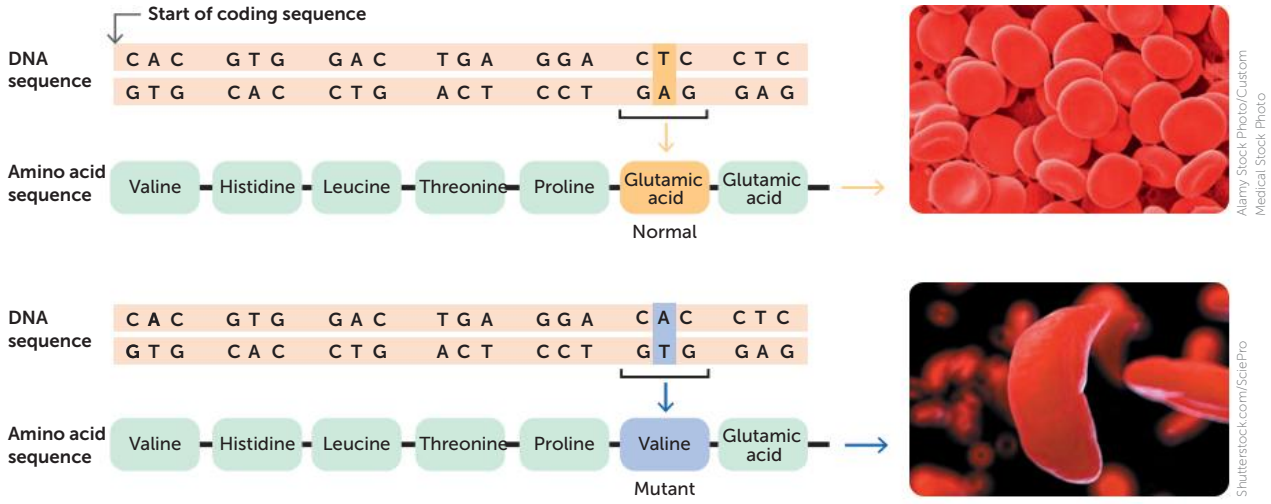

FIGURE 9.23 The point mutation of a substitution of one base pair results in sickle-cell anaemia

The sickle-shaped red blood cells often die early, resulting in **anaemia**. They are also inflexible and can become stuck in the blood vessels, causing a blockage. Other complications of sickle-cell anaemia include fatigue, jaundice, organ damage (such as to the kidneys, lungs and brain), high blood pressure and heart failure.

If a person with sickle-cell anaemia dies before reproducing, the allele that causes the disease is not passed on to the next generation. Therefore, you would expect that over many generations the frequency of the sickle-cell allele would gradually decrease until it was eliminated from the population altogether. On the other hand, if the ratio of mutation of normal alleles to sickle-cell alleles was great enough, it could cancel out the loss of alleles through the death of affected individuals. However, this is not the case: investigations have shown that the rate of alleles being lost from the population is about 100 times greater than the average rate of mutation at any point along a human chromosome. Some other mechanism must be at work to maintain the sickle-cell allele in the population. Figure 9.24 shows places in the world where the sickle-cell allele occurs in the population. When this is compared with Figure 9.21, you can see that the sickle-cell allele occurs only in areas where malaria is prevalent.

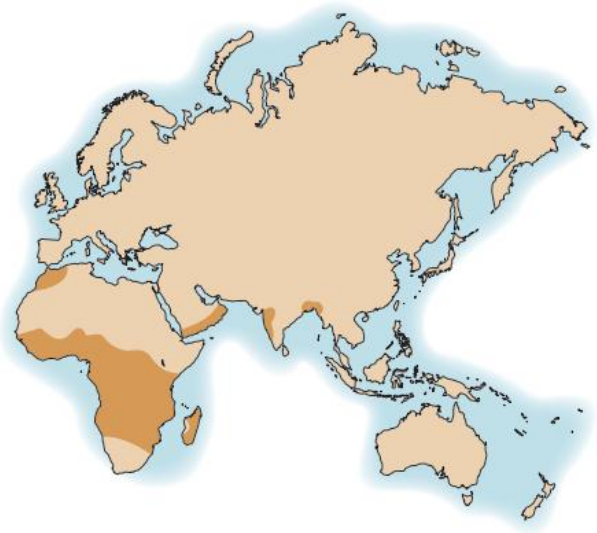

FIGURE 9.24 The distribution of sickle-cell anaemia

Anthony Allison was one of the first to notice the relationship between sickle-cell anaemia and malaria. He reported his observations in the *British Medical Journal* in 1954, noting that the sickling allele tended to have its highest frequency in areas where the risk from malarial parasites was greatest. He suggested that individuals with one sickle-cell allele were more resistant to malaria than those with normal haemoglobin in their red blood cells. This conclusion was based on Allison's observations that malarial patients who were also 'sicklers' had fewer malarial parasites than did malarial patients who were 'non-sicklers'. To gather evidence in support of these observations, Allison conducted a number of experiments. He inoculated both sicklers and non-sicklers with malaria and then treated those individuals in whom the disease developed. His results confirmed that the heterozygotes were less susceptible to infection from

malaria than individuals homozygous for normal haemoglobin. Further studies since Allison's pioneering experiments have supported his findings. It is now generally accepted that individuals heterozygous for the sickle-cell allele have a survival advantage in areas where malaria is prevalent.

The sickle-cell example shows how natural selection occurs in human populations. A mutation established a new allele in the population. Having one of these alleles gave individuals living in malarial-prone areas a survival advantage. This is known as a **heterozygote advantage**. Individuals with two sickle-cell alleles, those with sickle-cell anaemia, usually die. Those who are homozygous for normal haemoglobin are more susceptible to malaria. Therefore, the presence of malaria acted as a selective agent for the sickle-cell allele.

Activity 9.4
Investigating sickle-cell haemoglobin

Tay-Sachs disease

In section 9.1 you learnt that Tay-Sachs disease is a lethal condition caused by a mutation that results in the absence of the enzyme beta-hexosaminidase. This condition leads to a deterioration of the nervous system, and death usually occurs at a young age.

Tay-Sachs disease is a recessive condition found only in individuals who are homozygous recessive. People who are heterozygous have a reduced amount of beta-hexosaminidase. It appears that these individuals have some protection from tuberculosis, an infectious disease that primarily affects the lungs. This means that people who are:

- homozygous recessive die before reproducing, due to Tay-Sachs, and therefore do not pass on the recessive allele
- heterozygous survive tuberculosis, reproduce and therefore pass on both alleles
- homozygous dominant are affected by tuberculosis and may die prior to reproducing.

The heterozygote advantage provided by a heterozygous genotype increases the percentage of the recessive allele in the gene pools in areas affected by tuberculosis.

Thalassemia

Haemoglobin is made up of four protein chains that fit together. Two of these are alpha globin chains and the other two are beta globin chains. You have already learnt that sickle-cell anaemia affects haemoglobin molecules. Another disorder that alters the structure of haemoglobin is thalassemia. There are two forms of thalassemia.

- Alpha thalassemia is due to a mutation in the HBA gene on chromosome 16. This reduces the level of alpha globin in haemoglobin.
- Beta thalassemia is due to a mutation in the HBB gene on chromosome 11. This reduces the level of beta globin in haemoglobin.

Both of these conditions are inherited in an autosomal recessive manner. People with thalassemia have less haemoglobin in their red blood cells and, therefore, cannot carry as much oxygen in their blood. The severity of the disorder varies, depending on the number of affected genes, ranging from mild anaemia and fatigue to an enlarged liver and heart.

Thalassemia is more common in areas affected by malaria. Alpha thalassemia is more prevalent in South-east Asia, while beta thalassemia is more prevalent in the Mediterranean basin. Research has shown that malaria can act as a selective agent, resulting in an increased frequency of the alleles of alpha thalassemia. It is thought that the lower amount of haemoglobin gives some protection against malaria. Patients also seem to recover more quickly than those without thalassemia, possibly due to the increased number of red blood cells. It is possible that this is also the case with beta thalassemia, however, this has not been supported by research. Therefore, other factors may also influence the allele frequency. These include migration, genetic drift and founder effect.

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Questions 9.3

RECALL KNOWLEDGE

1 Define 'natural selection'.
2 Describe the phenotype of individuals who have each of the following genotypes for sickle-cell anaemia:
   a homozygous b heterozygous.
3 List the steps involved in evolution through natural selection.
4 Describe what a selective agent is, and give an example.

APPLY KNOWLEDGE

5 Explain why variation is crucial for natural selection.
6 Use a flow chart to summarise the history of our understanding of natural selection.
7 Describe how natural selection may have led to an increased occurrence of Tay-Sachs disease in areas where tuberculosis occurs.
8 Suggest the selective agent for each of the following characteristics.
   a Bacteria becomes resistant to antibiotics.
   b Inuit people tend to be short limbed and long bodied.
   c Prickly pear cacti have thorns on their flesh.
9 Explain how malaria can lead to an increased frequency of the allele for sickle-cell anaemia.
10 Use a Venn diagram to compare and contrast alpha thalassemia and sickle-cell anaemia.

9.4 GENETIC DRIFT

In any generation there is always the chance that some individuals will be more, or less, likely to pass on their alleles. This produces **genetic drift**, the random, non-directional change in allele frequency between generations. Genetic drift is not affected by whether an allele is beneficial or harmful; instead, it is purely by chance. Genetic drift occurs in populations of all sizes; however, it is unlikely to have a significant effect in large populations. It can, however, play an important role in evolution in small populations.

Genetic drift is also known as **random genetic drift** or the **Sewall Wright effect**, after the man who first recognised its significance in causing changes to allele frequencies. It is much the same as if you had 50 red balls and 50 black ones, with each ball able to reproduce itself periodically. If these 100 balls were placed in a bag and 50 balls were selected at random from it, the expected result would be 25 balls of each colour. It would not be surprising, though, to find that your sample contained 30 black balls and only 20 red ones. In this case, after

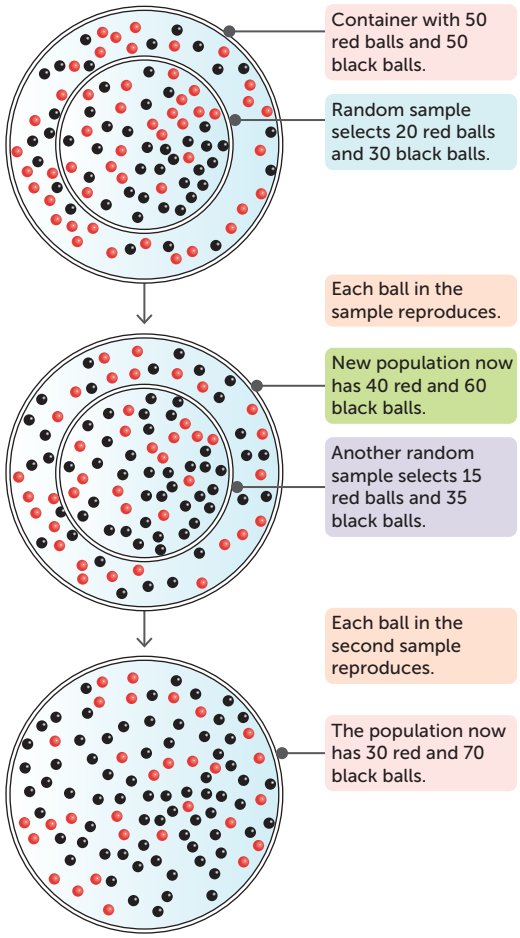

FIGURE 9.25
A model showing how random sampling of a pair of alleles can, by chance, increase the frequency of one allele and decrease the frequency of the other

reproducing, your new population would have 60 black balls and 40 red ones. If 50 balls were again selected at random, you would now expect the black ones to be favoured. This is how random genetic drift works in small human populations.

Studies have been carried out on isolated populations of Aboriginal Australians. One study investigated the isolated populations of Bentinck and Mornington Islands in the Gulf of Carpentaria. Originally these islands were part of the mainland, but rising sea levels cut them off and their populations became isolated. However, the Mornington Islanders maintained some contact with the mainland by using the smaller islands in between as 'stepping stones'. The blood group frequencies of the islanders have been studied and compared with those of the population occupying Bayley Point on the mainland. The occupants of Bentinck Island show allele frequency values for blood groups that fall outside the range for Aboriginal people in the rest of Australia. They show a very high proportion of the I^B allele and a complete absence of the I^A allele, unlike the mainland population, which has a low proportion of the I^B allele and a relatively high proportion of the I^A allele.

Key concept

Genetic drift is the random, non-directional change in allele frequency that occurs by chance. It is particularly significant in small populations.

Founder effect

An extreme example of genetic drift is the **founder effect**. This effect occurs when a small group moves away from its homeland to a totally new area and establishes a population, which later expands. Because of its small size, chance can cause the new groups to have:

- a different allele frequency from the original population
- decreased genetic variation.

This means that the new population may show a frequency of features that are not typical of the original homeland population.

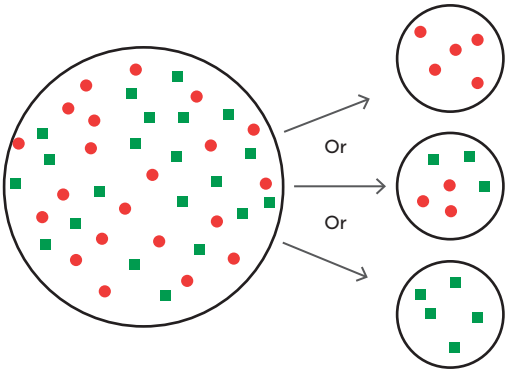

FIGURE 9.26
A model illustrating the founder effect. The original population is on the left and three different possible founding populations are on the right. It is possible for the founders to be quite unrepresentative of the original population. The chance selection of the founders will have a marked effect on the gene pool of later populations

Studies have been done on isolated groups to demonstrate this effect. One early and well-known study was by Bentley Glass and his co-workers in the 1950s on an isolated population in the United States. This group, known as the 'Dunkers', lives in Pennsylvania but originally came from Hesse, Germany. They are descended from Old German Baptist Brethren who came to the United States in the early 18th century. Their religion does not allow them to marry outside their group, and thus they constitute an isolated breeding population within the total population of the United States. The study investigated a number of easily measured physical traits, including the frequency of the ABO, Rh and MN blood groups, mid-digital hair, left- or right-handedness, and attached or free earlobes. For most of the traits studied, the Dunkers varied in allele frequency from the present-day population of Hesse and also from the surrounding American population. The environment for both the Dunkers and the surrounding American population is essentially the same, so there would not have been any natural selection to account for the differences in allele frequencies. Therefore, Bentley Glass concluded that genetic drift was responsible for this variation as the small size of the Dunker population allowed certain characteristics to become more common purely by chance.

Founder effect
This website provides more information about the founder effect.

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iStock.com/golibo

FIGURE 9.27
Left-handedness was one of the physical traits observed by Bentley Glass in his study of the Dunkers

Another example of the founder effect is the incidence of Tay-Sachs disease in the population of Ashkenazi Jews. Earlier in this chapter, Tay-Sachs disease was discussed as an example of a lethal recessive disease. Approximately 1 in 27 Ashkenazi Jews carries the altered allele, compared to 1 in 300 in non-Ashkenazi Jews. Ashkenazi Jews descended from a small number of individuals from Central or Eastern Europe. This group was initially isolated geographically. There was additional isolation through the custom of endogamy (only marrying within the community). This created a founder effect. The incidence of the mutated allele for Tay-Sachs disease was higher in the ancestors, and hence genetic drift is responsible for the prevalence of the disease in the current Ashkenazi Jew population.

The five most common Ashkenazi genetic diseases
This website has more information about the prevalence of disease in Ashkenazi Jews.

Genetic drift simulation
Use the simulation on this website to investigate the founder effect and the effect of a bottleneck.

Bottleneck effect

The **bottleneck effect** is another extreme example of genetic drift. In this situation, an event such as a natural disaster severely reduces the size of the population. The allele frequency after the disaster may, by chance, be different from before the event. It is important to note that, in the bottleneck effect, the chance of survival is by chance and not due to a specific trait.

An example of the bottleneck effect occurred in 1775 when a typhoon reduced the population of Pingelap, an island in Micronesia, to only 20. These survivors formed the founding population for the current inhabitants. Interestingly, among the survivors was a person heterozygous for achromatopsia. **Achromatopsia** is an inherited form of total colour blindness. The allele for achromatopsia is recessive. Today, after a number of generations, the incidence of achromatopsia on Pingelap is 5% of the population. In other parts of the world it is 0.0033%. Furthermore, 30% of the Pingelap population are carriers; they are not colour blind but they do have the affected allele. This is another example of how allele frequencies can change in small, atypical populations.

Key concept

The founder effect and bottleneck effect are two extreme examples of genetic drift that occur when a small group moves away from the main group or an event leaves only a small number of survivors.

Questions 9.4

RECALL KNOWLEDGE

1 Define:
   - **a** genetic drift
   - **b** founder effect
   - **c** bottleneck effect.

2 Explain how genetic drift is different from natural selection.

3 Use an example to explain how the founder effect may result in a population having different characteristics from another population of the same species.

APPLY KNOWLEDGE

4 Explain why genetic drift is unlikely to have a significant effect on the allele frequency of a large population.

5 Explain how it is possible that bushfires that affected areas of Victoria and New South Wales in early 2020 may reduce the genetic variation in koalas.

9.5 SPECIATION

All humans, from whatever part of the world and whatever ethnic background, have basic similarities and are capable of interbreeding to produce fertile offspring. That is, all humans belong to the same species. A **species** is a group of individuals that share many characteristics and are able to interbreed under natural conditions to produce fertile offspring.

Earlier in this chapter, isolation was mentioned as a barrier to gene flow. Reproductive isolation may lead to the development of separate gene pools. No two environments are exactly the same, so it would be expected that certain alleles would be favoured in one environment more than another. Therefore, over time the allele frequencies of each gene pool will change, depending on which characteristics are favoured for survival. Over many generations, the populations will become less and less alike as they develop characteristics that better suit them to their respective environments.

Speciation
This website provides an animation about the mechanisms of speciation.

If two populations are isolated for a very long period of time, and the environmental influences on each are different enough, major changes in the allele frequencies within each population could occur. In such a situation, the members of those populations may become so different that, even if the barriers to reproduction were removed, interbreeding would no longer be possible. If this occurred, the two populations would be regarded as separate species. The process of producing two species in this way is referred to as **speciation**.

The steps involved in speciation can be summarised as follows:

1 *Variation:* There is variation between individuals of a species.
2 *Isolation:* Populations of the same species are isolated without gene flow.
3 *Selection:* Each population is subjected to different selective agents.
4 *Speciation:* The allele frequency changes until they become so different that the two groups are no longer able to interbreed.

9.2 Evolutionary mechanisms

Key concept

Speciation, or the formation of new species, occurs due to variation, isolation and selection leading to two groups becoming so different that they can no longer interbreed.

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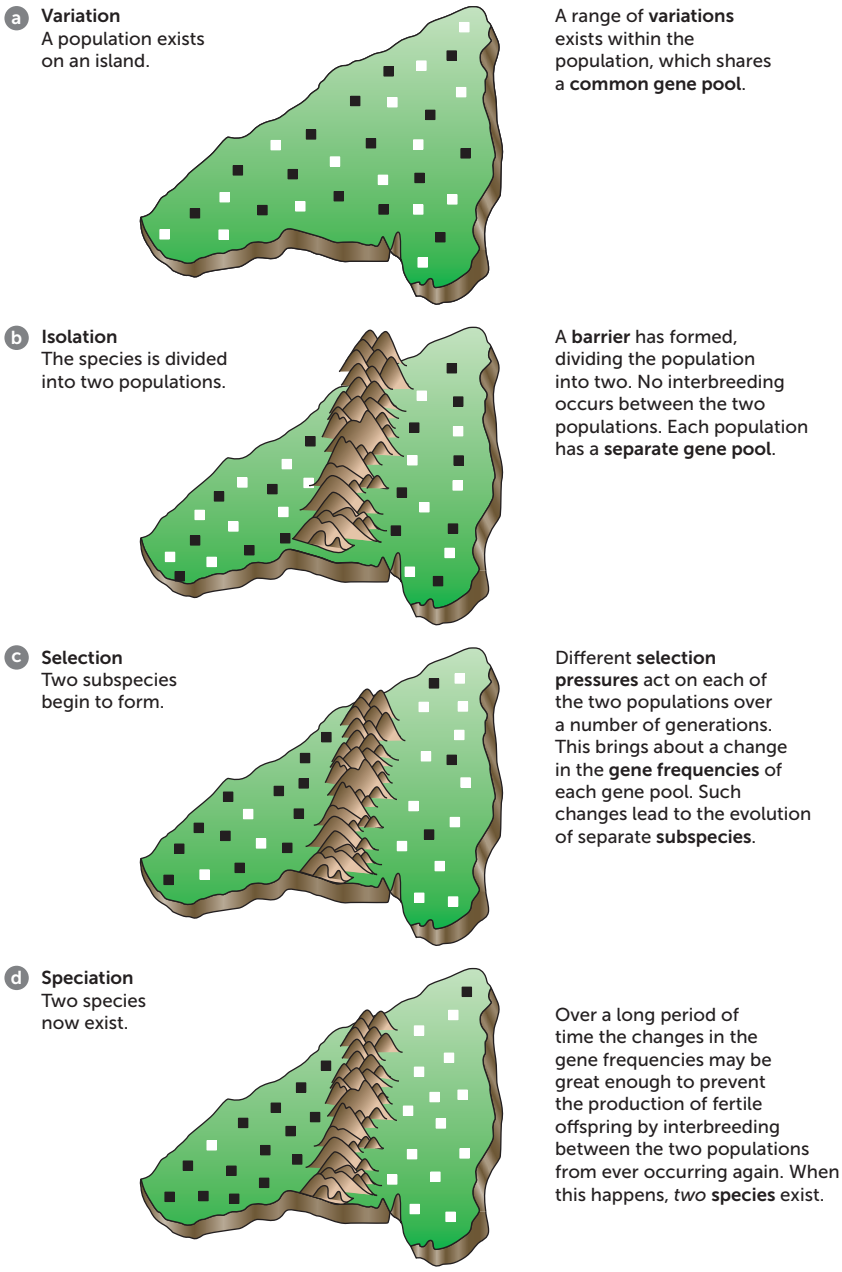

FIGURE 9.28
A diagrammatic representation of variation, isolation, selection and speciation

Questions 9.5

RECALL KNOWLEDGE

1 Define 'speciation'.
2 List the steps involved in speciation.
3 List three situations that may lead to isolation of groups.

APPLY KNOWLEDGE

4 Explain why variation is necessary for speciation.
5 Do you think it is likely that humans will form a new species in the future? Explain your answer.
6 Is mutation, natural selection or genetic drift the most important process in speciation? Justify your answer.
7 Would two groups of a species that are isolated in environments that are similar form new species? Explain why or why not.

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CHAPTER 9 ACTIVITIES

Developed by Southern Biological

ACTIVITY 9.1 Investigating the effect of ultraviolet radiation on *Saccharomyces cerevisiae*

We classify the broad spectrum of electromagnetic radiation from the sun into segments according to the effects we experience. For example, the warm sensation of sunshine on our skin is caused by invisible infrared radiation with wavelengths ranging from 700 nm to 1 000 000 nm (1 mm). Visible light is composed of wavelengths of between 400 nm (violet) and 700 nm (red). Radiation with wavelengths shorter than 400 nm but longer than 10 nm is classified as ultraviolet (UV) radiation. Radiation with wavelengths shorter than 10 nm is classified as X-rays. Some exposure to UV radiation is necessary for humans to produce vitamin D, but a careful balance is required because X-rays and UV radiation are destructive to many biological molecules, including DNA. Fortunately, the earth's atmosphere acts as a protective screen and filters out almost all the sun's radiation with wavelengths shorter than 290 nm. Nevertheless, the narrow UV band from 290 nm to 400 nm that can penetrate the atmosphere and reach the surface of the earth is capable of causing photochemical damage to DNA that can lead to skin cancer, so it is important to avoid over-exposure. As a defence against too much UV exposure, most organisms that are subject to the sun's rays have evolved to incorporate some level of DNA repair in their cell mechanisms. This confers a limited amount of inherent UV resistance.

Aim

To determine how ultraviolet radiation can be destructive for many biological molecules
Time requirement: 50 minutes

You will need

UV-sensitive yeast *Saccharomyces cerevisiae* starter plate; wild-type yeast starter plate; 8 sterile swabs; 8 YED agar plates; 4 plastic pipettes; 2 sterile culture tubes; Bunsen burner; permanent marker; sterile water; 2 sterile inoculation loops; ethanol or bleach; disposable gloves

Risks

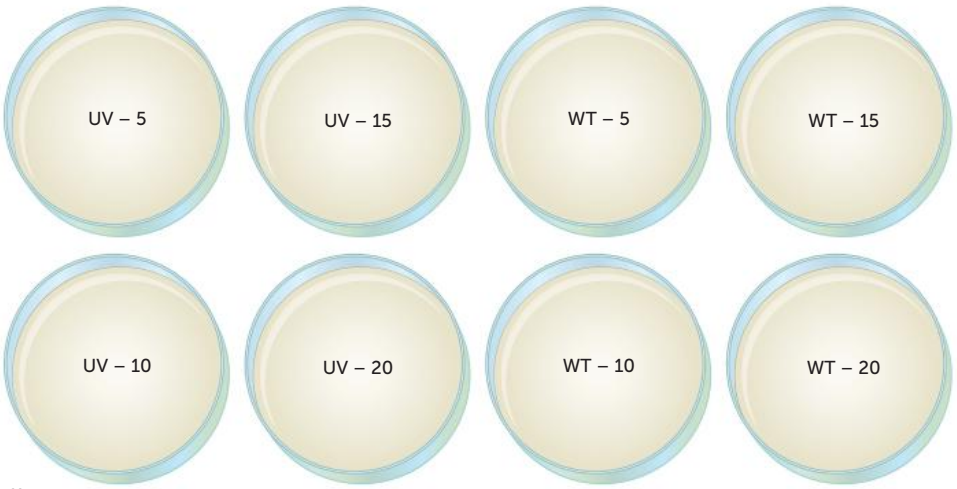
WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
While lab strains are usually harmless, fungi may cause disease, so assume them to be pathogenic.	Wear lab coats, safety glasses and gloves; wash hands thoroughly at end. Decontaminate benches before and after activity. Flood spills with bleach.
Micro-organisms will grow on the agar plates.	Do not open plates once they are securely taped. Dispose of plates appropriately after autoclaving.
Disposable gloves may pose an allergy risk.	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.

What to do

Note: To use aseptic technique, wipe your bench down with ethanol (or bleach) and keep your work near the Bunsen burner to take advantage of the updraught the flame will create to waft potential contaminants away from your materials.

Inoculation of exposure plates

1 Collect eight YED agar plates and label them as follows using a permanent marker.



Key:
UV = UV-sensitive yeast (mutated strain)
WT = wild-type yeast
Number = Time plate will be exposed to sunlight

2 Using a plastic pipette, add 1 mL of sterile water into a sterile culture tube.
3 Using a sterile inoculation loop, carefully scrape a single colony of the UV-sensitive yeast from the starter plate.
4 Select a large colony (>4 mm in diameter) or, if the colonies are small, scrape up two, or even three, on to the loop.
5 Place the loop in the water in the sterile tube and spin/swirl it to transfer the yeast into the sterile water.
6 Visually check that the cell mass has transferred from the loop to the sterile water.
7 Using a plastic pipette, immediately pump the liquid to distribute and suspend the yeast cells in the water. To do this, draw the liquid in and out of the pipette by squeezing and releasing the bulb of the pipette. Avoid introducing air bubbles or splashing the liquid up the sides of the tube. When finished, hold the tube up to the light to check that there are no visible lumps or particles in the water.
8 Dip a sterile swab into the yeast suspension and, as you withdraw it, press it against the sides of the tube to squeeze out excess water. It should come out moist but not dripping.
9 Using aseptic technique (see note above), 'swab' the surface of a YED plate in three directions to inoculate for a lawn culture, which is a culture that covers the entire plate as evenly as possible.
10 Immediately cover the plate to shield it from light and allow it to rest (right way up—that is, with the agar at the bottom) for a period of at least 15 minutes and up to one hour. This allows the moisture from the swab to be absorbed by the agar.
11 Repeat steps 8 to 10 for the remaining three UV-sensitive yeast plates. Then repeat this procedure for the four plates using the wild-type yeast.

Exposure to sunlight

1. State your hypothesis.
2. After the post-inoculation resting period, expose the one inoculated plate from each strain to direct sunlight for 5, 10, 15 and 20 minutes, respectively.
3. Immediately after exposure, incubate the plates in darkness for 48 hours at 30°C or 4 days at room temperature. For best results, follow these guidelines:
   - Keep the plate shielded from light until the last moment.
   - Use adhesive tape to attach the lid of the Petri dish to the base, but do not allow the tape to extend on to the surface of the lid where it will absorb UV light and shield the yeast from exposure.
   - Orient the plate so the lid is pointing directly at the sun. Aim to minimise the size of the shadow. If the sun's rays strike the lid at a glancing angle, most of the UV light will be reflected and the effectiveness of the exposure will be reduced.
   - Schedule the investigation at a time of year when you can be sure of bright, sunny conditions.
4. After the incubation period, observe and compare the level of coverage between the plates. Record your results in the table below.

Studying your results

1. Copy and complete the table below with the results of your experiment. Use the key below to indicate the level of coverage of the yeast on each agar plate.

+++	High coverage
++	Medium coverage
+	Low coverage
-	No coverage

UV exposure results

EXPOSURE TIME (MINUTES)	UV-SENSITIVE YEAST COVERAGE	WILD-TYPE YEAST COVERAGE
0		
5		
10		
15		
20		

2. Compare the results of your UV-sensitive yeast sample with the wild-type yeast sample. What differences do you observe? What conclusions can you draw from this data?
3. Graph your results.

Discussion

1. What is your independent variable?
2. What is the range of your independent variable?
3. What is your dependent variable?
4. What are your control variables and how did you control them?
5. What type of mutation does the UV-sensitive yeast portray?
6. Compare your results with others in your class. Were the results consistent?
7. Did your experiment support or refute your hypothesis, or were your results inconclusive?
8. Based on your findings, how does UV light impact the two different yeast strains? Do they differ? If they do, explain why.
9. Suggest how your findings might relate to evolution.

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Taking it further

To protect our skin from harmful UV rays, we apply different sunscreens with different sun protection factor (SPF) values. Do these values have any merit, and do commercially produced sunscreens offer better protection than alternatives such as coconut oil, clothing material and sunglass lenses?

ACTIVITY 9.2 Venusians: Investigating natural selection

Venusians are an imaginary group of people from the planet Venus. Because of the intense heat, their skin is jet-black; all individuals are homozygous for skin colour. If a mutation occurs resulting in a Venusian of a lighter skin colour, the individual usually dies before being able to reproduce. However, one such mutation created a brown-skinned individual who did survive and reproduced, passing the new allele to some of his children. The skin of the individuals affected by this mutant allele was extremely thick, providing them with added resistance to a lethal biting insect.

As time went by, the number of Venusians with the mutant allele increased. However, when two of these individuals produced children, homozygotes died in infancy.

In this activity, we will investigate how the mutant allele becomes distributed through the population over time. To simplify our activity, we will start with heterozygotes, and assume that all those who are homozygous for the mutant allele die before they can reproduce. We will also assume that one out of every three Venusians who are homozygous for the normal allele dies from a lethal insect bite.

You will need (for each pair)

Two containers: 2 L ice-cream containers work well; 20 black beads or counters to simulate the black skin allele (B) in each gamete; 20 white beads or counters to simulate the brown skin allele (b) in each gamete; felt pen; tally sheet; pencil

What to do

1. Label one container 'Male Parent' and the other 'Female Parent'.
2. Place 10 of the black beads in each container, then add 10 of the white beads.
3. Prepare a tally sheet similar to the one below using the symbol 'B' for the black skin allele and 'b' for the brown skin allele.

	GENOTYPES IN THE VENUSIAN OFFSPRING		
	BB	Bb	bb
1			
2			
3			
4			
5			
6			
7			
8			
9			
10			

4. Simulate reproduction by shaking the containers well and drawing out one bead from each. The beads represent gametes. Use the colour of the beads to determine the genotypes of the offspring. Place a tick in the relevant box on the tally sheet, then replace the beads.
5. When you have completed 10 draws, place the beads back into the containers. Your partner should repeat steps 1 to 4 above. Together you should have two completed tally sheets.

Studying your data

1. Because the first column contains individuals that are homozygous for black skin ('BB'), only two out of every three survive to adulthood. Tally the number of offspring that will survive to produce the next generation.
2. Individuals with the genotype 'bb' will all die in the first year. If you eliminate these individuals, how many surviving offspring do you now have?
3. What is the ratio of black skin to brown skin offspring that survive to adulthood?
4. Combine your data with the other groups in your class to obtain a bigger sample. What is the ratio now?

Interpreting your data

1. How has this activity shown that mutations that increase an individual's chances of survival and reproduction affect the proportions of particular characteristics in a population?
2. What has happened to the proportion of the allele 'b' in the population? Has it been entirely eliminated? Do you think it ever will be?
3. Summarise how this chance mutation has helped the survival of the Venusian population.

ACTIVITY 9.3 Modelling natural selection

In this activity, you will model the effects of natural selection on a hypothetical population of frogs. The frogs are naturally green, yellow or orange in colour, and are preyed upon by water birds. You will simulate the different predation rates on the three variations of frog colour over a number of generations by throwing a die.

You will need (for each group)

Coloured cards or counters – 30 green, 30 yellow and 30 orange; a die

What to do

1. Draw up a table similar to the following one to record your results.

Number of different coloured frogs in successive generations

GENERATION NUMBER	GREEN FROGS	YELLOW FROGS	ORANGE FROGS
1	10	10	10
2			
3			
4			
5			
6			
7			
8			
9			
10			

2. From the pool of coloured cards, select 10 of each colour. These will be your first generation of frogs. Shuffle these cards so that they are well sorted, and then deal them out in pairs. You should have 15 pairs of cards representing 15 pairs of frogs.

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3 We will assume that each pair of frogs consists of a male and a female, and that each pair produces only one offspring. The pairs produce offspring according to the following rules.
   - Two green frogs produce a green offspring.
   - Two yellow frogs produce a yellow offspring.
   - A green frog and a yellow frog produce an orange offspring.
   - Two orange frogs produce a colour that can be decided by the throw of a die:
     1 = a green offspring
     2 = a yellow offspring
     3 or 4 = an orange offspring
     5 or 6 – throw the die again until you get a 1, 2, 3 or 4.
   - A green frog and an orange frog produce a colour that can be decided by the throw of a die:
     1, 2 or 3 = a green offspring
     4, 5 or 6 = an orange offspring.
   - A yellow frog and an orange frog produce a colour that can be decided by the throw of a die:
     1, 2 or 3 = yellow
     4, 5 or 6 = orange.
4 Because their colours do not blend into the background so easily, yellow and orange frogs are more likely to be preyed upon by birds than are the green frogs. Simulate predation in your population of 45 frogs. Fifteen of the frogs are to be taken as prey. Throw a die 15 times and for each throw remove one frog according to the following rules.
   - If 1, 2, 3 is thrown, remove a yellow frog.
   - If 4 or 5, remove an orange frog.
   - If 6, remove a green frog.
5 You should have 30 cards remaining. This is your second generation of frogs. Count the cards and record the number of each colour in the table.
6 Shuffle the cards well and repeat steps 3, 4 and 5 to get the third generation. Record your results in the table.
7 Continue the process until all the frogs are the one colour, or until you have completed 10 generations.

Studying your results

1 Which colour frog became the most frequent in the population? Why do you think this was the case?
2 Which colour frog was eliminated first? Explain why this occurred.
3 Compare your results with other groups in the class. Have all groups obtained similar results? How much variation was there in the results between the different groups?

Interpreting your results

1 How has this activity modelled the process of natural selection? In your answer, describe what was creating the selection pressure on the population of frogs.
2 Explain why there was variability, if any, between the groups in your class.
3 What changes would you have to make to predation by the water birds to achieve a completely orange population of frogs? Repeat the activity with your changed parameters. Was your prediction correct?
4 Over several generations, what would happen to the composition of the frog population if water birds preyed equally on the three frog colours?
5 Write a summarising paragraph, using the principles of natural selection, to link the breeding patterns of the frogs and predation by water birds.

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ACTIVITY 9.4 Investigating sickle-cell haemoglobin

In an article published in the *British Medical Journal* in 1954, AC Allison first put forward the hypothesis that the possession of the sickle-cell allele may have a selective advantage in areas where malaria is prevalent. In investigating this proposition, Allison inoculated 30 African adult male volunteers with malaria and then observed them for 40 days. At the end of the period of observation, he treated all the participants with a prolonged course of antimalarial chemotherapy.

Allison's volunteers were of similar age and none had been in an area where malaria occurred for at least 18 months. They all appeared to be comparable except for the presence or absence of the sickle-cell allele. His results are shown in the following table.

Allison's results from the inoculation of Africans with malaria

	NUMBER OF PARTICIPANTS	DEVELOPED MALARIA	DID NOT DEVELOP MALARIA
With a sickle-cell allele	15	2	13
Lacking a sickle-cell allele	15	14	1

Interpreting the results

1. What was Allison's dependent variable? What was his independent variable?
2. What factors did Allison appear to control in his experiment?
3. Which group of subjects was the control group, and which the experimental?
4. Did Allison's results support his hypothesis? Explain why you think so.
5. Do the results Allison obtained suggest a reason why the sickle-cell allele has survived in Africa?
6. Refer to Figures 9.21 and 9.24. Does the information provided in these figures support your answer to Question 5? Give reasons for your answer.
7. Explain how the high incidence of the sickle-cell allele in parts of Africa could be considered an example of natural selection.
8. Would a university ethics committee today be likely to approve an experiment such as the one that Allison performed? Give reasons for your answer.

Allison's article describing his investigation

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CHAPTER 9 SUMMARY

- Evolution is the change in characteristics of a species over time due to changes in allele frequencies. It is influenced by mutations, migration, natural selection and genetic drift.
- A population is a group of organisms of the same species living in the same location at the same time. Geneticists study the frequency of alleles in the gene pools of populations. Variations in allele frequencies reflect differences in characteristics.
- Mutations are changes in the DNA that can occur spontaneously or due to exposure to a mutagen such as ionising radiation. They may affect a single gene (gene mutation) or more than one gene (chromosomal mutation).
- Mutations may be classified based on the following:
  - *Cause:* They can be spontaneous or induced.
  - *Heritability:* Mutations in somatic cells are not inherited (somatic mutations), but mutations in gametes are inherited (germline mutations).
  - *Effect:* Missense mutations change the protein produced, nonsense mutations produce a shorter protein that is not functional, neutral mutations change an amino acid but not the functioning of the protein, while a silent mutation does not change the amino acids and so the protein remains the same.
  - *Extent:* Gene mutations only affect one gene, while chromosomal mutations affect a number of genes or the whole chromosome.
  - *Change in DNA:* A nucleotide may be inserted, substituted or deleted; sections of DNA may be duplicated, deleted or translocated; chromosome pairs may not separate during meiosis.
- Insertion or deletions of nucleotides will result in a frameshift unless they are of a multiple of three nucleotides. This means that the base codes are read from a different base and, therefore, all the amino acids from that point on are affected.
- Examples of conditions due to mutations are: Duchenne muscular dystrophy and cystic fibrosis, due to gene mutations; Down syndrome, Patau syndrome and Klinefelter syndrome, due to trisomy; and Cri-du-chat syndrome and Turner syndrome, due to monosomy.
- Tay-Sachs disease is a recessive, autosomal lethal condition due to a mutation in the HEXA gene where the enzyme needed to break down GM2 ganglioside isn't produced. This results in a build-up of the toxic fatty substance that destroys neurons. Babies show intellectual and physical deterioration from age three to six months, and die early in childhood.
- Gene flow is the movement from one population to another. It is enabled by migration and stopped by some barriers such as geographical barriers, religion, language and sociocultural barriers.
- The theory of evolution by natural selection was proposed by Charles Darwin and Alfred Russel Wallace. It was based on the observation that there is variation within a species, the birth rate is greater than food supplies can sustain, and the species' numbers remained relatively constant. This led to the idea that species struggle for existence and to the theory of survival of the fittest. This means that favourable characteristics led to survival and were reproduced in the process of natural selection. The environmental factor that determines the survival is called the selective agent.
- Evolution occurs by natural selection, when the alleles for the favourable characteristics are passed on to future generations and therefore increase in frequency. This changes the characteristics of the species over a number of generations (evolution).

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- Sickle-cell anaemia is due to a point mutation on the HBB gene which changes the structure of haemoglobin, resulting in red blood cells with a distorted shape that become crescent shaped in low oxygen levels. The homozygous form is often fatal; however, the heterozygous form has milder symptoms and actually gives the individual protection from malaria. Therefore, in malaria-affected areas, the frequency of the sickle-cell allele is higher as it gives a survival advantage.
- Tay-Sachs disease is a fatal, recessive condition; therefore, affected individuals do not live long enough to reproduce. People who are heterozygotes have a selective advantage due to being protected from tuberculosis. Therefore, they will survive and reproduce, passing on the recessive and dominant alleles. This means that the incidence of Tay-Sachs disease is higher in areas affected by tuberculosis.
- Thalassemia is a gene mutation that results in changes to either the alpha globin or beta globin proteins in haemoglobin. Individuals with thalassemia have more red blood cells with less haemoglobin than unaffected individuals. These appear to give them some protection against malaria. Therefore, the incidence of thalassemia is greater in malaria-affected areas.
- Genetic drift is the random, non-directional change in allele frequency. It occurs in all populations; however, it usually only has a significant effect on small populations. It occurs when, by pure chance, more of one allele is passed on than others.
- The founder effect is an extreme example of genetic drift that occurs when a small group moves away from the original population. If, by chance, the allele frequency of the new group differs from the original population, then the frequency of features will also be different.
- The frequency of Tay-Sachs disease is greater in the population of Ashkenazi Jews than in other populations. This is due to a higher incidence in the common ancestor and to the fact that the Ashkenazi Jews are isolated by traditionally marrying within the religion.
- The bottleneck effect occurs when the population size is dramatically reduced by something like a natural disaster. The allele frequency of the remaining individuals may, by chance, be different from the original population.
- A species is a group of individuals that share many characteristics and are able to interbreed to produce fertile offspring. Evolution may lead to new species when the allele frequencies change so much that they are no longer able to interbreed. The process of producing new species is called speciation.
- Speciation occurs due to different selective pressures applied to different groups of a species. It goes through the process of variation, isolation, selection and speciation.

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CHAPTER 9 GLOSSARY

Achromatopsia An inherited form of total colour blindness

Albinism An inherited inability to produce pigment in hair, skin and eyes

Allele frequency How often each allele of a gene occurs in a population

Anaemia A condition in which there is a reduced amount of haemoglobin in the blood, or a reduced number of red blood cells

Aneuploidy A change in the chromosome number as a result of non-disjunction

Bottleneck effect An extreme form of genetic drift that occurs when the size of a population is severely reduced due to a sudden event such as a natural disaster. The allele frequency of survivors may not reflect that of the original population

Chromosomal mutation A change to the structure and/or number of chromosomes in an organism

Cri-du-chat syndrome A rare genetic disorder caused by a missing part of chromosome 5

Cystic fibrosis A disorder controlled by a recessive allele carried on an autosome that is incurable but can be detected during foetal development; mucus-secreting glands, particularly in the lungs and pancreas, become fibrous and produce abnormally thick mucus, resulting in, among other things, chest infections

Down syndrome *see* trisomy 21

Duchenne muscular dystrophy A genetic disease resulting in wasting of leg muscles and then arms, shoulders and chest

Evolution The gradual change in the characteristics of a species

Evolved Having gone through the process of evolution

Founder effect A type of genetic drift where a new population is formed by a small number of individuals; the small sample size can cause marked deviations in allele frequencies from the original population

Frameshift A mutation involving an insertion or a deletion that results in a change in the way that the sequence is read

Gene flow The transfer of alleles from one population to another through migration

Gene mutation An alteration to a single gene

Gene pool The sum of all the alleles carried by the members of a population

Genetic drift *see* random genetic drift

Geneticist A scientist who specialises in the study of genetics

Genotype The combination of alleles for a gene

Geographical barrier A feature of the landscape that prevents populations from interbreeding; includes oceans, mountain ranges, large lake systems, deserts and expansive ice sheets

Germinal mutation *see* germline mutation

Germline mutation A change in the hereditary material in the egg or sperm that becomes incorporated into the DNA of every cell in the body of the offspring

Heterozygote advantage A situation where a heterozygous genotype has a higher chance of survival than either homozygous genotype

Induced mutation A mutation caused by a mutagenic agent

Klinefelter syndrome A genetic disorder resulting from inheritance of two X chromosomes and one Y chromosome

Lethal recessive A recessive allele that, inherited in the homozygous condition, results in the death of the embryo, foetus or child

Migration The movement of people from one area to another with the intention of settling permanently

Missense mutation A mutation that causes a change in an amino acid resulting in a different protein being produced

Monosomy Where an individual has only one copy of a chromosome instead of two

Mutagen *see* mutagenic agent

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Mutagenic agent An environmental agent that increases the rate of mutation

Mutant An organism with a characteristic resulting from a mutation

Mutation A change in a gene or chromosome leading to new characteristics in an organism

Natural selection The process by which a species becomes better adapted to its environment; those individuals with favourable characteristics have a survival advantage and so pass those characteristics on to subsequent generations

Neutral mutation A mutation that causes a change in an amino acid; however, it does not cause an overall change in the protein

Nonsense mutation A mutation that results in a STOP codon, producing a shortened peptide chain

Partial monosomy Where part of a pair of chromosomes is missing

Patau syndrome A genetic disorder resulting from an extra copy of chromosome 13

Phenotype The observable characteristic due to the genotype

Phenylketonuria (PKU) An inherited disease resulting in damage to the growing brain and, thus, extreme intellectual deficiency, a tendency towards epileptic seizures, and failure to produce normal skin pigmentation

Point mutation A change in just one of the bases in a DNA molecule

Population A group of organisms of the same species living together in a particular place at a particular time

Random genetic drift The occurrence of characteristics in a population as a result of chance rather than natural selection; occurs only in small populations; also called genetic drift or Sewall Wright effect

Selective agent Any factor that causes the death of organisms with certain characteristics, but which has no effect on individuals without those characteristics

Sewall Wright effect *see* random genetic drift

Sickle-cell anaemia An inherited disease causing early death; results from the inheritance of two alleles for sickle-cell anaemia

Silent mutation A mutation that does not change the sequence of amino acids

Sociocultural barrier Barrier to interbreeding that is due to social or cultural factors

Somatic mutation A change occurring in a gene in a body cell (not in a gamete)

Special creation The belief that a god created all species

Speciation The process of new species developing

Species The basic unit of biological classification; members of a species are capable of interbreeding and producing fertile offspring

Spontaneous mutation A mutation that occurs due to an error in a natural biological process

Struggle for existence A principle where the number of organisms is greater than the resources in the environment can support; therefore, there is competition between the organisms for these resources

Survival of the fittest A principle whereby organisms with favourable characteristics survive, but organisms with unfavourable characteristics die before they have a chance to reproduce

Tay-Sachs disease (TSD) A genetic disorder caused by a missing enzyme that results in fatty substances accumulating in the nervous system

Trisomy 21 A genetic disorder resulting from an extra copy of chromosome 21 or an extra part of chromosome 21; also called Down syndrome

Turner syndrome A genetic disorder resulting from inheritance of one X chromosome and no other sex chromosome

Variation The differences that exist between individuals or populations of a species

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CHAPTER 9 REVIEW QUESTIONS

Recall

1 Define a 'population'.

2 What do scientists mean when they speak of a 'gene pool'?

3 a Define 'mutation'.
  b List the ways that the DNA may be changed in a mutation.
  c Distinguish between gene mutations and chromosomal mutations.
  d Give an example of a congenital disorder that can be caused by a gene mutation and one that can be caused by a chromosomal mutation.

4 a What are mutagens (or mutagenic agents)?
  b List five examples of mutagenic agents.

5 What is a lethal recessive?

6 a Distinguish between trisomy and monosomy.
  b Give an example of each condition.

7 Briefly describe the significance of the founder effect in human evolution.

8 a Define 'gene flow'.
  b List the common barriers that may lead to the isolation of one gene pool from another, and give examples of each type.
  c List five different kinds of sociocultural barriers to gene flow, and describe how each is thought to act.

9 Outline the main points of Darwin's theory of natural selection. Include an explanation of the terms 'struggle for existence' and 'survival of the fittest'.

Explain

10 Explain the difference between somatic and germline mutations.

11 Explain how mutations could change the proportion of certain alleles in a gene pool.

12 a Explain what random genetic drift is.
  b Select a modern population in which genetic drift is thought to have had an effect and describe why this might be the case.

13 Using the example of Tay-Sachs disease, explain how genetic diseases can lead to changes in allele frequencies in a population.

14 People of short stature tend to live in cold climates, and people with long limbs and short torsos tend to live in hot climates. Explain how these adaptations to cold and hot environments could have come about.

15 a What is sickle-cell anaemia?
  b Explain why sickle-cell anaemia is usually lethal.
  c List the advantages and disadvantages of having the sickle-cell trait in an area where malaria is prevalent.

16 How could isolation lead to selection and speciation?

Apply

17 Why does special care need to be taken when pregnant women require X-rays?

18 Summarise the pattern of inheritance that occurs in genetic disorders such as Duchenne muscular dystrophy. When there is no history of such disorders in a family, how are they thought to arise?

19 Discuss why mutations occurring in the reproductive cells are considered more important than those occurring in the body cells. In your discussion, describe the possible long-term effects of the two situations.

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20 The more often cells divide, the greater the risk of errors and mutations. For this reason, scientists have hypothesised that when a baby is born with a congenital disorder caused by an error in cell division, the father is the parent more likely to have contributed the gene with the mutation. Compare the number of eggs produced by a female with the number of sperm produced by a male and explain why scientists have proposed this hypothesis.

21 Lethal recessive alleles result in the death of an individual. How would this affect the allelic composition of the gene pool?

22 The risk of having a baby with Down syndrome increases as the mother gets older. The following table shows the relationship between Down syndrome and maternal age.

a Draw an appropriate graph to display the data in the table.

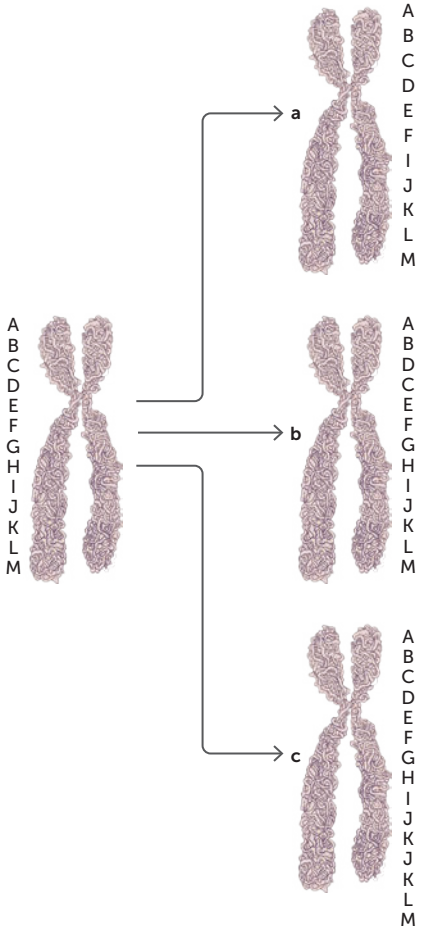
b The risk of a baby having any chromosome abnormality increases dramatically with increasing maternal age. Suggest reasons for this.

Mother's age and risk of having a baby with Down syndrome

AGE OF MOTHER (YEARS)	RISK OF DOWN SYNDROME
20	1 in 1667
23	1 in 1429
26	1 in 1176
29	1 in 1000
32	1 in 769
37	1 in 227
40	1 in 106
43	1 in 50
46	1 in 23
48	1 in 14
49	1 in 11

Source of data: Adapted from Dr Mark Hill 2020, UNSW Embryology

23 The following figure shows the sequence of the genes A to M on a chromosome. What type of chromosomal mutation is represented by each of a, b and c?



24 During the 14th century, plague epidemics drastically reduced the human population of Europe. Use this as an example to describe the way natural selection operates so that only the fittest tend to survive.

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25 According to a recent report, 13% of Scotland's population are redheads. Two out of every five Scots carry the allele for red hair. However, only 2% of the world's population are estimated to be natural redheads.

- **a** Suggest a reason for the high frequency of the allele for red hair in the gene pool of the Scots.
- **b** In the population of Scotland, what do you think will happen to the frequency of the allele for red hair over time? Give reasons for your answer.

26 A team of American scientists has been trying to develop a vaccine to give permanent immunity against malaria. What do you think will happen to the frequency of the sickle-cell gene within a population if this vaccine is effective? In writing your answer, ensure that you explain the adaptive value of the various genotypes and the selection pressures on each.

Extend

27 Western Australia has been a world leader in the application of carrier detection to reduce the incidence of Duchenne muscular dystrophy.

- **a** What does carrier detection involve?
- **b** What takes place following the detection of a carrier?
- **c** What is preventing the complete elimination of Duchenne muscular dystrophy?

28 Malthus claimed that species of organisms always produce more offspring than the existing resources can support. Is this true of the human species in the past or at present? Is it likely to be true of the human species in the future?

29 Describe the barriers to gene flow that exist for the following populations:

- **a** groups in South Africa
- **b** groups in the islands of Polynesia, such as New Zealand, Tahiti and Hawaii
- **c** Jewish people.

30 Using analysis of mitochondrial DNA, researchers have determined that all humans are descended from a woman who lived in Africa 200 000 years ago – the so-called mitochondrial Eve. If we are all descended from a common ancestor, how is it that there are so many different types of humans today? Describe the processes that must have taken place to produce the differences between present-day groups of humans.

31 Speculate on what might be the long-term effect on allele frequencies if a mutation suddenly produced a favourable allele that gave a natural resistance to all forms of heart disease.

32 One of the best-researched investigations into natural selection is the work of a British geneticist, Henry Bernard Davis Kettlewell, on the peppered moth, *Biston betularia*. The peppered moth gets its name from the scattered dark markings on its otherwise pale wings and body. The moth flies at night and rests by day on tree trunks. These trunks are usually encrusted with lichens, and the pale-coloured moth is practically invisible against this background.

However, in 1849, a coal-black mutant form of the moth was found near Manchester in England. Within a century, this black form had increased to 90% of the population in this region. The change in allele frequency that occurred in this example is a good model of how natural selection takes place.

Find out:

- **a** how the black form of the moth became the more prevalent variant
- **b** which form is the most prevalent today.

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10 BIOTECHNOLOGY PROVIDES EVIDENCE OF EVOLUTION

UNIT 4 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including the use of virtual or real biotechnological techniques of polymerase chain reaction (PCR), gel electrophoresis for deoxyribonucleic acid (DNA) sequencing, and techniques for relative and absolute dating, safely, competently and methodically for valid and reliable collection of data
- represent data in meaningful and useful ways; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and sample size may influence uncertainty and limitations in data; and select, synthesise and use evidence to make and justify conclusions
- select, use and/or construct appropriate representations, including phylogenetic trees, to communicate conceptual understanding, solve problems and make predictions

SCIENCE AS A HUMAN ENDEAVOUR

- developments in biotechnology have increased access to genetic information of species, populations and individuals, existing now or in the past, the interpretation and use of which may be open to ethical considerations
- developments in the fields of comparative genomics, comparative biochemistry and bioinformatics have enabled identification of further evidence for evolutionary relationships, which help refine existing models and theories

SCIENCE UNDERSTANDING

Evidence for evolution

- biotechnological techniques provide evidence for evolution by using PCR (to amplify minute samples of DNA to testable amounts), bacterial enzymes and gel electrophoresis to facilitate DNA sequencing of genomes
- comparative studies of DNA (genomic and mitochondrial), proteins and anatomy provide additional evidence for evolution; genomic information enables the construction of phylogenetic trees showing evolutionary relationships between groups

Source: School Curriculum and Standards Authority, Government of Western Australia

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The great majority of scientists accept the general idea of evolution. However, this idea has always been controversial and there are still many people who, for a variety of reasons, do not accept the idea that species evolve. It is important, therefore, to look carefully at the evidence available to support the theory of evolution. Besides the study of fossils, much of the evidence for evolution has come from comparative studies. Traditionally, the focus for comparative studies has been on anatomy and embryology, but the development of technology in more recent times now allows comparative studies to be conducted on both proteins and DNA.

DNA also helps us to understand why evolution happens. When Darwin proposed the theory of evolution, he based it on evidence gained through observation. We now understand the structure of DNA and the inheritance process. This allows us to make sense of the evidence provided and understand the changes that led to them.

10.1 PROCESSING DNA

DNA can be processed by a range of techniques so that it can be analysed and compared for applications such as tracking evolutionary changes. In this section, we will be focusing on:

- making many copies of the sample using polymerase chain reaction (PCR)
- cutting the DNA into smaller lengths with restriction enzymes from bacteria
- separating the lengths of DNA with gel electrophoresis to produce a DNA profile
- determining the sequence of nucleotides, or their bases, using Sanger's method.

Polymerase chain reaction and amplifying DNA

One of the early limitations of DNA analysis was the amount of DNA needed. In many cases there was insufficient DNA, or the organism may have been adversely affected in order to collect enough. The development of the **polymerase chain reaction (PCR)** has enabled small quantities of DNA to be replicated, producing testable amounts to use in analysis techniques. Kary Mullis was awarded the Nobel Prize in Chemistry in 1993 in recognition of his development of the PCR technique.

Nobel prizes
This website provides the press release for the 1993 Nobel Prize in Chemistry.

PCR mimics the natural process of DNA replication that occurs prior to cell division. During the process, the DNA goes through a series of three steps:

1. *Denaturing:* The two strands of DNA are separated.
2. *Annealing:* Short sections of DNA (primers) are bound to the separated strands.
3. *Extension:* The short sections of DNA are extended to produce longer strands.

This sequence is repeated approximately 20–30 times, in a process called **thermocycling**. It takes two to three hours to produce about a billion copies of the DNA.

Denaturation

During natural DNA replication, the enzyme helicase separates the two strands of DNA, allowing each strand to be copied. The polymerase chain reaction uses heat to achieve the same function. Temperatures of approximately 94–96°C are used to break the hydrogen bonds holding the two strands together. This separates the strands without disrupting each individual strand.

Annealing

During the annealing process the temperature is decreased to approximately 50–60°C. This allows short strands of DNA called **primers** to bind to the single DNA strands. The primers are not random sections of DNA. Instead, they are complementary to either end of the section of DNA to be copied.

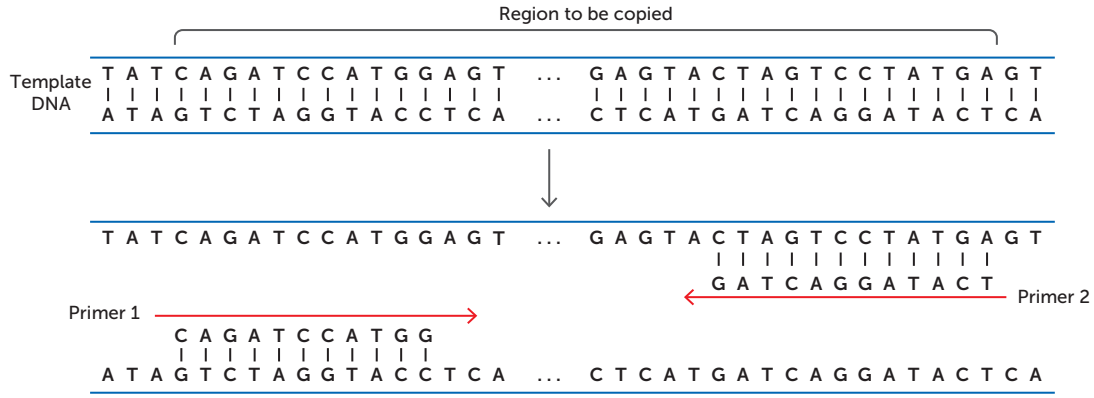

FIGURE 10.1 Primers that are complementary to either end of the region to be copied are used during the annealing process

Extension

The extension step, also known as elongation, also mimics the process of DNA replication. The enzyme DNA polymerase is used to join new, complementary nucleotides to the sections originating with the primers. This extends, or elongates, the nucleotide chain and creates a new strand of DNA. However, it is not the full length of the original DNA, as it starts at the primer and not at the end of the DNA. Eventually, the majority of DNA strands are the length of DNA between the location of the primers.

DNA polymerase attaches to double-stranded DNA. Prior to the extension step, this only occurs where the primers are located. Therefore, the primers act as a starting point and, hence, initiate DNA replication.

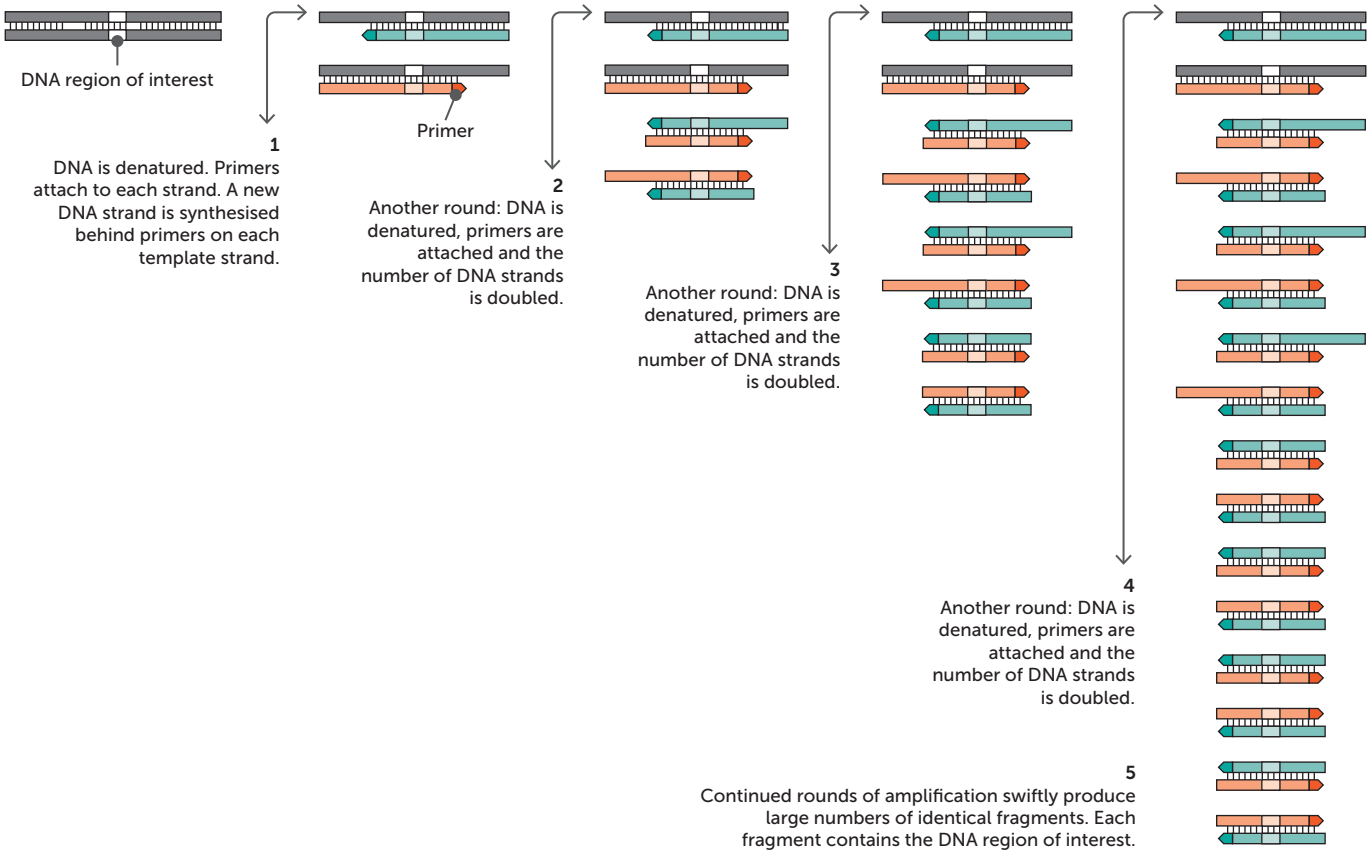

FIGURE 10.2 A diagrammatic representation of the polymerase chain reaction

In the PCR process, the denaturation step is carried out at 94–96°C. However, at this temperature, the DNA polymerase is usually destroyed. This means that more of the enzyme has to be added after the heating stage of each cycle. This is very time consuming and uses large amounts of DNA polymerase.

Therefore, almost all PCR applications now use a heat-stable DNA polymerase. One such enzyme, taken from a heat-loving bacterium called *Thermus aquaticus*, is called Taq polymerase. It does not denature when heated and has allowed the procedure to be simplified and automated, permitting the PCR sample to be alternately heated and cooled. Taq polymerase's optimal temperature is 68–72°C; therefore, the extension phase is carried out at this temperature.

PCR
This website provides an animated sequence on PCR.

PCR virtual laboratory
This website provides a PCR virtual laboratory.

Key concept

The polymerase chain reaction uses heat, primers and DNA polymerase to amplify a section of DNA, producing many copies from a small amount.

Gel electrophoresis and DNA profiling

In Chapter 8, you learnt about **restriction enzymes** and their ability to cut DNA at specific nucleotide sequences. These enzymes can be used in a range of applications, including DNA analysis. When restriction enzymes are added to DNA, it cuts the strands into different lengths depending on the base sequence of the specific DNA sample. The length of these pieces can be analysed and compared with other DNA samples.

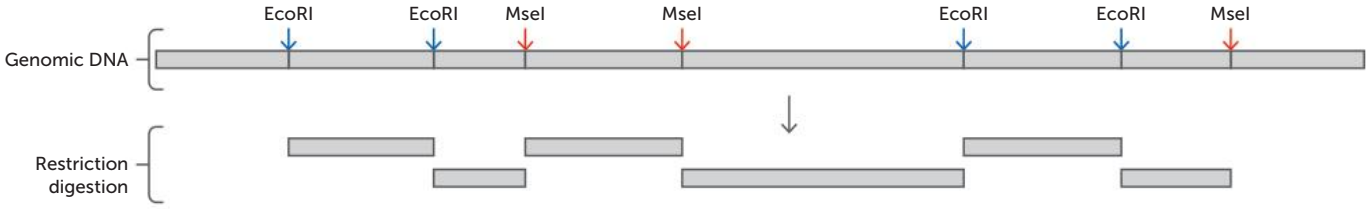

FIGURE 10.3 DNA cut with the restriction enzymes EcoRI and MseI producing pieces of different lengths

Gel electrophoresis is a technique that is able to separate DNA strands based on their lengths. The DNA pieces are placed in **wells** in a semi-solid gel that is immersed in a solution of an electrolyte. There are electrodes at either end of the gel. The negative electrode is closest to the DNA and the positive electrode is at the opposite side. When an electric current is passed through the gel, the negatively charged DNA moves towards the positive electrode. The smaller DNA pieces move faster than the larger ones and so are located further away from the negative electrode when the current is stopped. This results in a pattern of bands that looks similar to the barcodes on products sold in supermarkets. This banding pattern is an individual's **DNA profile**, often called a **DNA fingerprint**.

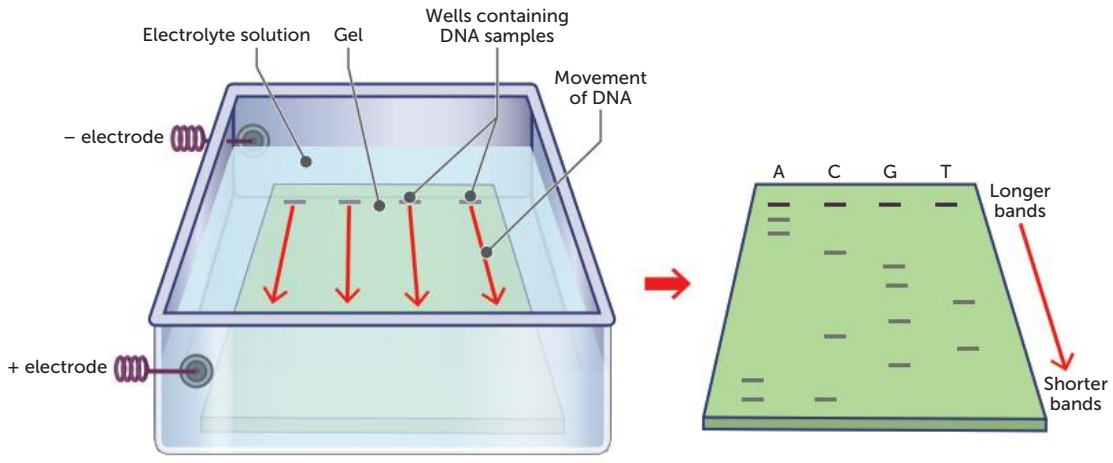

FIGURE 10.4
An illustration of the apparatus commonly used for gel electrophoresis and the resulting profile

Gel electrophoresis
This website contains more information about gel electrophoresis.

Filling the wells

Using a micropipette
This website has more information about how to use a micropipette.

The wells where the DNA is placed are simply depressions in the gel. This means that, when the current is applied, the DNA will move through the gel rather than diffuse through the solution. Therefore, for gel electrophoresis to work effectively the DNA needs to be accurately placed in the wells. This is typically done using a **micropipette**. Micropipettes have disposable tips at the end that can be put on and off the pipette without any contact, reducing any chance of cross-contamination.

iStock.com/Eakkalak Khiwsuwannasri

FIGURE 10.5 Micropipette

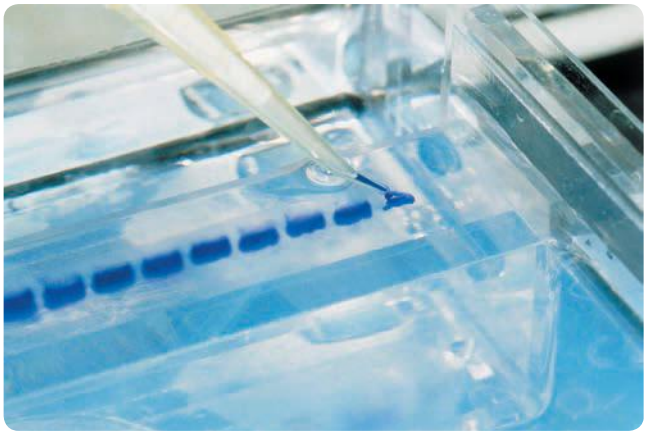

FIGURE 10.6 Micropipettes are used to place the DNA samples into the wells of the gel

DNA ladders

A DNA ladder is often 'run' at the same time as the samples. The ladder contains segments of DNA with known lengths. The results from the unknown sample are compared to the ladder to determine the length of the DNA strands in the sample.

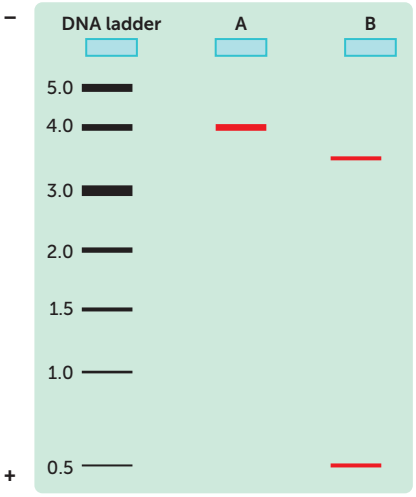

FIGURE 10.7 Samples of DNA can be compared to the DNA ladder

Visualising DNA

Activity 10.1
Investigating electrophoresis simulation

After the DNA strands are separated, scientists use different methods to visualise the DNA. Ethidium bromide can be added to the agar prior to the gel being set. As the DNA moves through the gel it picks up some of the chemical. Upon completion of the 'run', a special ultraviolet light is shone over the gel and the DNA fluoresce. Unfortunately, ethidium bromide is a carcinogen and, therefore, must be handled very carefully.

Activity 10.2
Investigating the effect of restriction digestion enzymes on lambda DNA

Methylene blue is a dye that binds to DNA. When the gel is soaked in the dye, the areas containing DNA stain a deeper blue and are therefore visible to the naked eye. Another method is the use of DNA probes. These are short sections of a single strand of DNA with a radioactive or fluorescent molecule that binds to the DNA being tested.

Key concept

A DNA profile is produced by using restriction enzymes to cut DNA into smaller lengths that are separated by gel electrophoresis, which uses an electrical current to move the DNA segments through the gel at a rate proportional to their length.

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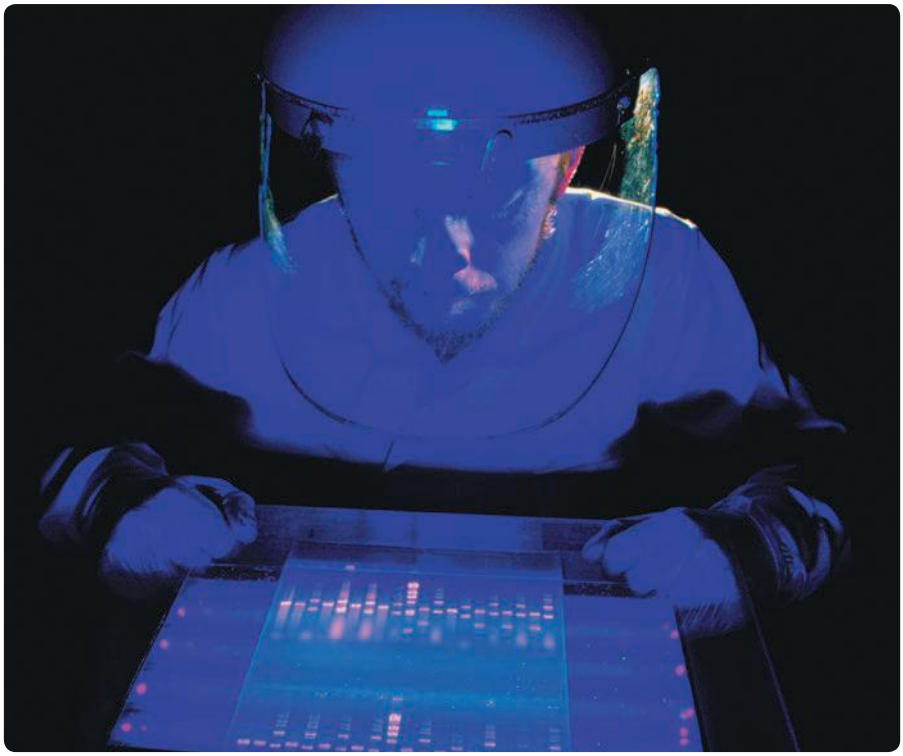


Alamy Stock Photo/Science Photo Library

FIGURE 10.8
Gel electrophoresis stained with ethidium bromide and placed under UV light

DNA sequencing

DNA sequencing is the determination of the precise order of nucleotides in a sample of DNA. The method most frequently used to determine such a sequence was invented by Frederick Sanger, who was awarded his second Nobel Prize in Chemistry in 1980 for this accomplishment.

DNA is synthesised from four nucleotides, each with a different nitrogenous base – adenine, cytosine, guanine or thymine. Nucleotides are more correctly called deoxynucleotide triphosphates, as they consist of three phosphate groups joined to the sugar deoxyribose with its base.

When DNA forms:

- each nucleotide loses two phosphate groups
- the sugar molecule loses a hydrogen atom from the hydroxy group (OH) when it bonds to the phosphate group of an adjacent nucleotide.

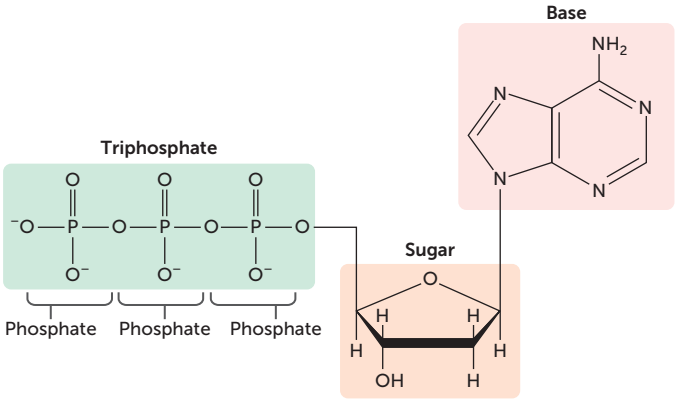
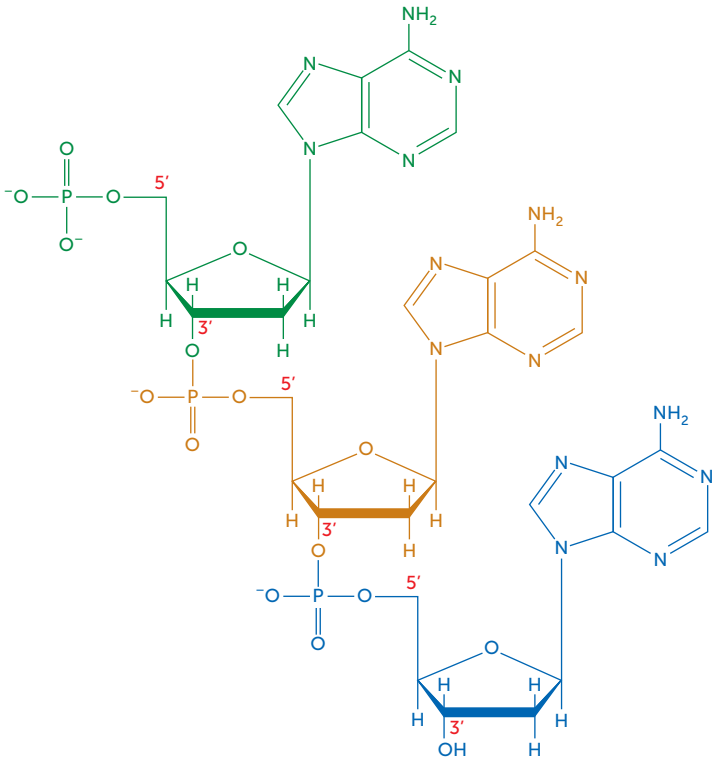


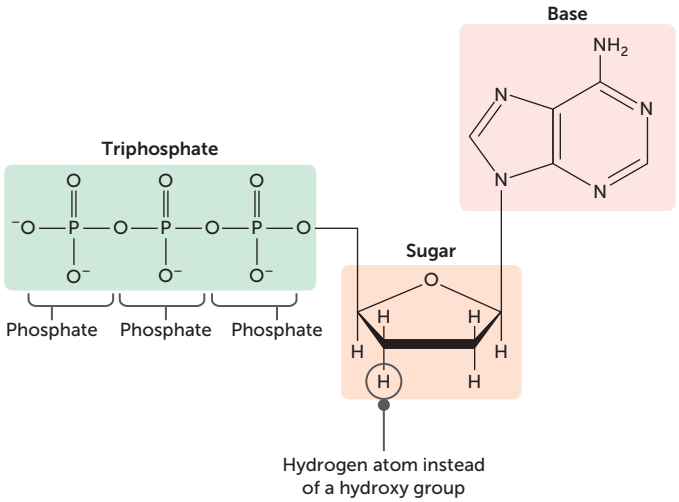
FIGURE 10.9
Structure of a nucleotide

FIGURE 10.10
Three nucleotides joined together in a section of DNA



In Sanger's method of determining a DNA sequence, synthetic nucleotides that lack the OH group are added to the growing strand. These are called dideoxyribonucleotide triphosphates, or **dideoxyribonucleotides** (ddNTPs).

FIGURE 10.11
Structure of a dideoxyribonucleotide triphosphate



The synthetic nucleotide stops the elongation of the sequence because there is no OH group for the next nucleotide to attach to. This happens at each of the nucleotides in the DNA sample, creating different lengths of DNA. These can be separated using gel electrophoresis. Knowing which base was added to create each length allows scientists to determine the order of nucleotides.

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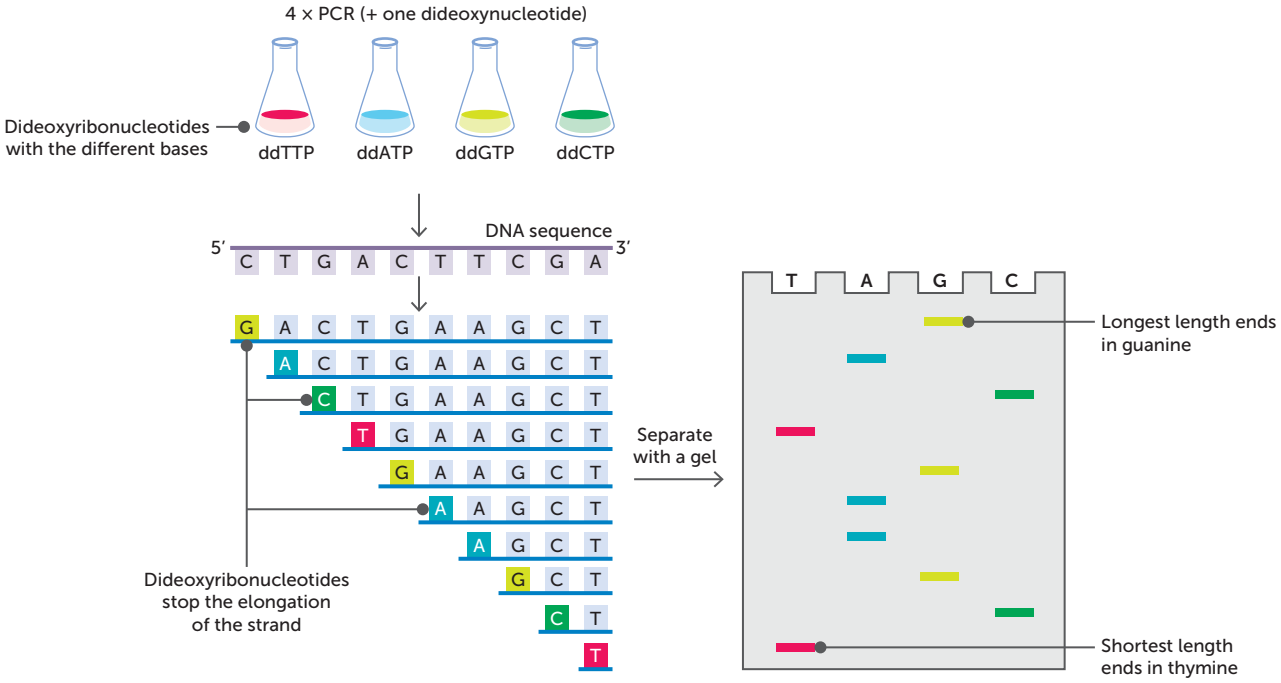
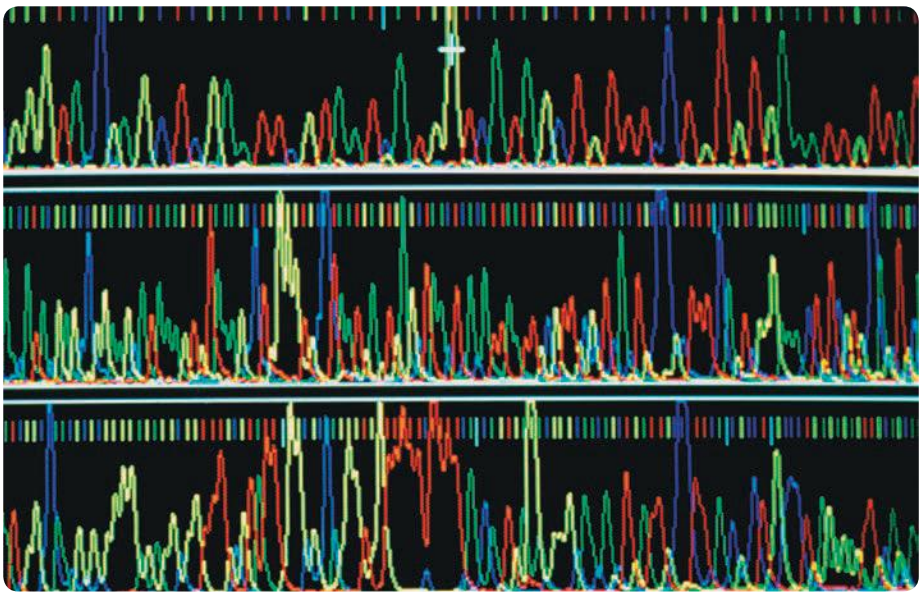


FIGURE 10.12 DNA sequencing using Sanger's method

DNA sequencing can be used to identify mutations or to compare DNA from different organisms. This is useful in identifying inherited disorders such as sickle-cell anaemia, cystic fibrosis and some forms of cancer. It has also been used for maternity and paternity tests, in cases where the identity of the father or mother of a child is in dispute. DNA sequencing can be used by scientists to compare species in order to track evolutionary changes.

Sanger sequencing
This website shows an animation of the Sanger method of DNA sequencing.

DNA sequencing
This website provides a series of annotated slides on DNA sequencing.



Alamy Stock Photo/Science Photo Library

FIGURE 10.13 A DNA sequence displayed on a computer screen

Key concept

The sequence of nucleotides in DNA can be determined by methods such as Sanger's method, which uses dideoxyribonucleotides to stop the lengthening of the DNA strand. This produces segments of different lengths that can be separated by gel electrophoresis.

Activity 10.3
Investigating biotechnological techniques

Ethical considerations of genetic information
This website contains more detailed information about the ethical considerations of genetic information.

Ethical considerations with genetic information

As technologies continue to advance, it is becoming easier and cheaper to obtain genetic information. As with the use of all technologies, the ethical use of genetic information is an important consideration. This includes respecting the following principles.

- *Autonomy*: respect for the right to be self-determining and to choose whether or not to be tested and, if tested, to know and share the information. It also includes the right of an individual to decide their own future, independent of genetic information.
- *Confidentiality*: the use of genetic information is treated sensitively, and is accessed only by those who are authorised to access it.
- *Equity*: the right to fair and equal treatment regardless of genetic information.
- *Privacy*: the right to be 'left alone' and to make decisions regarding genetic testing and the resulting information, independent of others.

key concept

Ethical factors such as autonomy, confidentiality, equity and privacy need to be considered when gaining and using genetic information.

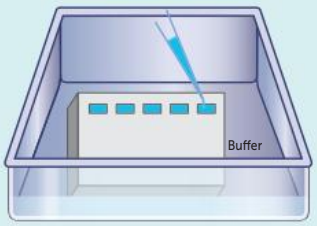
Questions 10.1

RECALL KNOWLEDGE

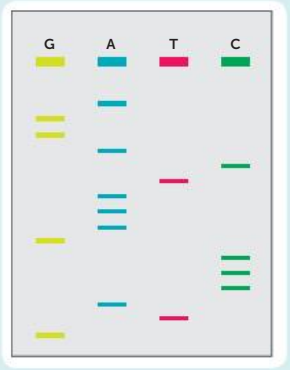
1 What does 'PCR' stand for?
2 List the three steps in PCR.
3 Explain the role of a primer in PCR.
4 What is the advantage of using Taq polymerase over other DNA polymerases?
5 Explain the role of restriction enzymes in DNA profiling.
6 Will the shorter or longer lengths of DNA travel the greatest distance during electrophoresis? Explain your answer.
7 Name the instrument used to place the DNA in the wells for gel electrophoresis.
8 Explain why a DNA ladder is useful in interpreting results from electrophoresis.
9 List three methods of visualising DNA after electrophoresis.
10 Draw a simplified structure of:
   a deoxynucleotide triphosphate
   b dideoxyribonucleotide triphosphate.

APPLY KNOWLEDGE

11 Explain why temperatures of approximately 96°C are sufficient to denature the DNA.
12 Suggest what would happen if the temperature were too high during the annealing step of PCR.
13 'DNA sequencing makes it possible for suitable primers to be chosen for PCR.' Discuss this statement.
14 The image below shows DNA being put into the wells of an agarose gel prior to electrophoresis. Label the positive and negative sides and explain why it must be placed this way.



15 The diagram below shows the bands produced from electrophoresis during DNA sequencing using Sanger's method. Write the base sequence for the section of DNA.



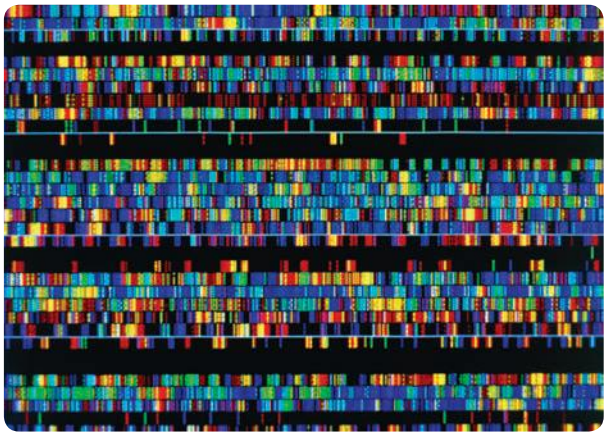
16 Identify two situations when it may be unethical to use genetic information. For each situation, discuss the reasons for and against its use.

10.2 DNA PROVIDES EVIDENCE OF EVOLUTION

Information obtained in DNA profiles and DNA sequencing can be used to compare the DNA of different organisms.

Comparative genomics

Scientists have determined that all living things use the same DNA code, adding weight to the hypothesis that all living things are related to each other and have evolved from a common ancestor. Although all species of organisms have DNA, the sequence of bases in the DNA varies. New genes are gained by mutation; others are lost by natural selection, genetic drift or some other process. Despite the common ancestor, the code in the DNA is different for different species.



Science Photo Library/JAMES KING-HOLMES

FIGURE 10.14 Computer screen display of the human DNA sequence. Each colour represents a specific base. The sequence of bases makes up the genetic code

When speciation occurs, the new species would have very similar DNA. However, as the new species gradually change through mutations, natural selection and genetic drift, they accumulate more and more differences in their DNA. Species that are more distantly related have more differences in their DNA, whereas species that are more closely related share a greater portion of their DNA.

The complete set of DNA in each cell of an organism is called the **genome**. **Comparative genomics** is a relatively new field of biological research in which the genome sequences of different species are compared. By comparing the sequence of the human genome with genomes of other organisms, researchers are able to identify regions of similarity and difference. This procedure provides an effective means of studying evolutionary changes among organisms, helping to identify genes that are preserved among species, as well as genes that give each organism its unique characteristics.

TABLE 10.1 Comparative genome sizes of a number of organisms

ORGANISM	ESTIMATED SIZE (BASE PAIRS)	CHROMOSOME NUMBER	ESTIMATED GENE NUMBER
Human	3.0 billion	46	21 000
Mouse	2.9 billion	40	21 000
Fruit fly	165 million	8	13 000
Roundworm	97 million	12	19 000
Yeast	12 million	31	6 000
Bacteria	4.6 million	1	3 200

The successful completion of the Human Genome Project in 2003 demonstrated that major sequencing projects can generate high-quality data. Consequently, interest in sequencing the genomes of many other species rose significantly. By analysing the genomic features that have been preserved in a number of species over millions of years, researchers are beginning to tease apart the often-subtle differences between animal species. Comparative genomics has revealed a high level of similarity between closely related organisms such as humans and chimpanzees. It has also been used to reveal the diversity of gene composition in different evolutionary lineages. Such research may result in a rearrangement of the way we view some of the evolutionary relationships between primates.

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10.1 Biotechnology

DNA and evolution
This website provides a light-hearted account of using DNA sequencing.

Examination of the genome of our closest living relatives, the chimpanzees, shows that they share more than 98% of our DNA (Table 10.2). Scientists quote slightly higher or lower figures depending on what exactly is being compared. Interestingly, humans have 23 pairs of chromosomes, while chimpanzees have 24 pairs. Scientists believe that, at some time in the past, two small chromosomes found in chimpanzees fused to form one of the human chromosomes.

TABLE 10.2 Relationship between humans and great apes using DNA differences

PRIMATES BEING COMPARED	DNA DIFFERENCE (%)
Human–chimpanzee	1.2
Chimpanzee–gorilla	1.2
Human–gorilla	1.6
Chimpanzee–orangutan	1.8
Human–orangutan	2.4
Gorilla–orangutan	2.4

Endogenous retroviruses

In addition to the genes coding for proteins, chromosomes also contain some non-coding sequences of bases in the DNA. These sequences are sometimes referred to as 'junk DNA' as they have no apparent function and appear to serve no purpose. Comparisons of junk DNA provide similar results as those for other parts of the genome: more closely related species have more junk sequences in common. This observation only makes sense if related species have evolved from a common ancestor.

Good examples of stretches of apparently non-functional DNA are **endogenous retroviruses (ERVs)**. An ERV is a viral sequence that has become part of an organism's genome. Retroviruses store their genetic information as RNA, not DNA. Upon entering a cell, a retrovirus copies its RNA genome into DNA in a process known as **reverse transcription**. The DNA then becomes inserted into one of the host cell's chromosomes. A retrovirus only becomes endogenous if it inserts into a cell whose chromosomes will be inherited by the next generation, an ovum or a sperm cell. The offspring of the infected individual will then have a copy of the ERV in the same place, in the same chromosome, in every single one of their cells. All subsequent generations will also have a copy of the ERV at the same location. What scientists have found is that ERVs make up 8% of the human genome, and that other primates also possess some of the same ERVs in exactly the same locations in their genomes.

For example, when comparing the chromosomes of humans and chimpanzees, it has been discovered that the same ERVs are located in the short arm of chromosome 10, the short arm of chromosome 1, the long arm of chromosome 9 and the short arm of chromosome 6 for both species. In all, scientists have found 16 instances of human ERVs matching exactly with chimpanzee ERVs. This is compelling evidence that humans and chimpanzees share a common ancestor. Any retrovirus that became inserted into the genome of a common ancestor would be inherited by both chimpanzees and humans at exactly the same location in the chromosome.

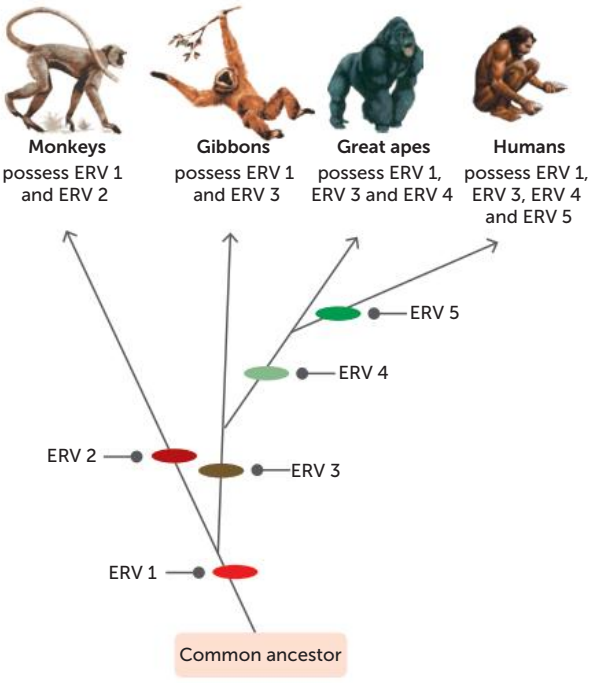

FIGURE 10.15 Simplified example of how endogenous retroviruses could be used to trace common ancestry: the great apes and humans have a more recent common ancestor as they share more endogenous retroviruses

Mitochondrial DNA

Mitochondria are the organelles in the cell where the aerobic phase of respiration occurs to release energy for use by the cell. Most of a cell's DNA is located in the nucleus, but a small amount is in the mitochondria. This is called **mitochondrial DNA**, or **mtDNA**.

Unlike the DNA in the nucleus, which is in the form of very long strands, mitochondrial DNA is in the form of small circular molecules. There are about five to ten of these molecules in each mitochondrion. Mitochondrial DNA has 37 genes, all of which are essential for the mitochondrion to function normally. Twenty-four of the genes contain the code for making transfer RNA molecules, which are involved in protein synthesis. The other 13 genes have instructions for making some of the enzymes necessary for the reactions of cellular respiration.

Most cells contain large numbers of mitochondria and therefore usually have between 500 and 1000 copies of the mtDNA molecule. This makes it a lot easier to find and extract than the DNA in the nucleus, and so smaller samples can be used. In humans, the mtDNA genome consists of about 16 500 base pairs, representing only a fraction of the total amount of DNA in a cell.

Inheritance of mitochondrial DNA

Human eggs and sperm both have mitochondria, but while an egg has many hundreds, a sperm has only about 100, just enough to provide the energy for the sperm to swim to the egg. After a sperm has penetrated the egg at fertilisation, the mitochondria in the sperm are rapidly destroyed. This means that, while our nuclear DNA comes from the nucleus of the egg and the sperm, our mitochondrial DNA comes only from the egg. In other words, we inherit nuclear DNA from both parents, but we inherit mitochondrial DNA only from our mothers. You got your mitochondrial DNA from your mother; she got it from her mother, and so on.

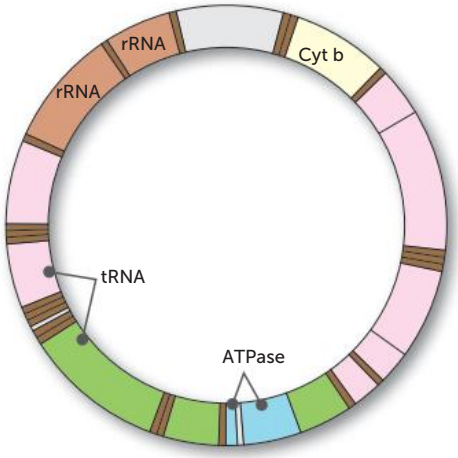

FIGURE 10.16 Model of a molecule of mtDNA showing the location of some of the genes

Evidence from mitochondrial DNA

DNA found in the mitochondria has a higher rate of mutation than nuclear DNA. Because of these mutations, human mtDNA has been slowly diverging from the mtDNA of our original female ancestor, and the amount of mutation is roughly proportional to the amount of time that has passed. Scientists are able to use the similarity between the mtDNA of any two individuals to provide an estimate of the closeness of their relationship through their maternal ancestors. If their mtDNA is identical they will be closely related, perhaps even siblings. On the other hand, if the mtDNA is very different, their last common maternal ancestor lived long ago. The use of mtDNA has been found to be of most value when comparing individuals within a species and for species that are closely related. In this way, it has allowed scientists to track the ancestry of many species back hundreds of generations. For example, through studying mitochondrial DNA it has been possible to trace the migration routes of ancient peoples. Such studies have shown that most Europeans are descended from hunter-gatherers who migrated into Europe during the last Ice Age, rather than from farmers coming from the Middle East. It has also been used to demonstrate the evolutionary relationships between humans and closely related species.

Analysis of mtDNA has become an important tool in mapping the relationships between species. Using such analysis, scientists can verify evidence of evolution gained from other sources. For example, examination of mtDNA has shown that the last common ancestor of modern humans and Neanderthals lived around 600 000 years ago.

mtDNA and Neanderthals
This website provides information about the identification of the genes in mitochondrial DNA of Neanderthals.

Questions 10.2

RECALL KNOWLEDGE

1. Where do scientists gain information from when comparing DNA?
2. Define 'genome'. Describe how sequencing the genome can be used to provide evidence for evolution.
3. Define 'endogenous retrovirus' and 'non-coding DNA'.
4. Describe how endogenous retroviruses are used as evidence for evolution.
5. What do the genes on mtDNA code for?
6. Why do we only inherit mitochondrial DNA from our mothers?

APPLY KNOWLEDGE

7. The DNA of dogs is 85% similar to that of humans, while the DNA of chimpanzees is 98% similar to that of humans. Explain how this information supports the idea that we have a more recent common ancestor with chimpanzees than with dogs.
8. Explain why not all retroviruses are endogenous retroviruses, and why only endogenous retroviruses are useful in providing evidence for evolution.
9. Explain why comparison of structures such as endogenous retroviruses and mitochondrial DNA was not available prior to the development of techniques such as electrophoresis and DNA sequencing.
10. Compare and contrast mitochondrial DNA and nuclear DNA.

10.3 OTHER BIOCHEMICAL EVIDENCE

DNA is not the only chemical substance that can provide evidence for evolution. Other techniques include a comparison of proteins such as cytochrome C and bioinformatics.

Protein sequences

Comparative protein studies also provide evidence for evolution. Proteins consist of long chains of particular amino acids linked together in a precise sequence determined by the DNA. There are tens of thousands of types of proteins in living things and all are fabricated from 20 kinds of amino acids.

Modern biochemical techniques enable the sequence of amino acids in a protein to be determined. By comparing the type and sequence of amino acids in similar proteins from different species, the degree of similarity can be established. Animals of the same species have identical amino acid sequences in their proteins, and those from different species have different amino acids or they are arranged in a different order. Just like DNA analysis, the degree of difference between proteins enables an estimate to be made of the amount of evolution that has taken place since two species developed from a common ancestor. The longer the period of time involved, the greater the number of amino acids that are different.

Amino acids are usually represented by a three-letter code, frequently the first three letters of their name. To make comparison of amino acid chains easier, scientists have also adopted a system of coding whereby one letter is used to represent one particular amino acid. By listing the amino acids for a particular protein in sequence, a comparison can be made with other species. This has been done for a number of proteins that appear to be in all species and which are referred to as **ubiquitous proteins**. Such proteins perform very basic, but essential, tasks that all organisms require for life. Ubiquitous proteins are found in all organisms, from bacteria to humans, and are completely independent of an organism's specific function or the environment in which it lives. Such proteins carry out the same functions no matter where they are found.

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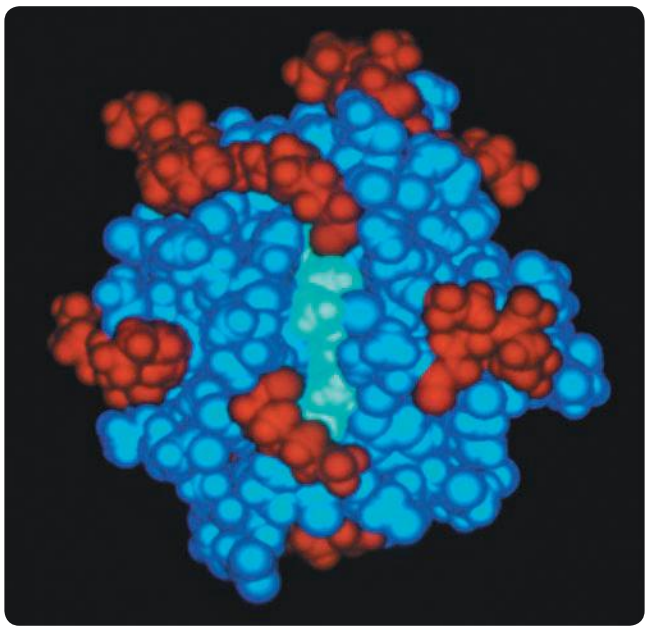
TABLE 10.3 The three-letter and single-letter codes used for amino acids

AMINO ACID	THREE-LETTER CODE	SINGLE-LETTER CODE
Alanine	Ala	A
Arginine	Arg	R
Asparagine	Asn	N
Aspartic acid	Asp	D
Cysteine	Cys	C
Glutamic acid	Glu	E
Glutamine	Gln	Q
Glycine	Gly	G
Histidine	His	H
Isoleucine	Ile	I
Leucine	Leu	L
Lysine	Lys	K
Methionine	Met	M
Phenylalanine	Phe	F
Proline	Pro	P
Serine	Ser	S
Threonine	Thr	T
Tryptophan	Trp	W
Tyrosine	Tyr	Y
Valine	Val	V

Cytochrome C is a well-researched example of a ubiquitous protein that shows how protein sequences can provide evidence for evolution. This protein performs an essential step in the production of cellular energy. It appears to have changed very little over millions of years of evolution. Human cytochrome C contains 104 amino acids. Regardless of the species tested, 37 of these have been found at the same positions in every sequenced cytochrome C molecule. This strongly suggests that these proteins have descended from an ancestral cytochrome C molecule found in a primitive microbe that existed more than 2000 million years ago.

To compare cytochrome C sequences, they need to be aligned so that the maximum number of positions containing the same amino acids can be determined. The more similarity there is between two molecules, the more recently they have evolved from a common ancestor. By doing such comparisons, scientists have determined that the cytochrome C of chimpanzees and gorillas is the same as that for humans, and for rhesus monkeys it differs by only one amino acid compared with that of humans.



Alamy Stock Photo/RGB Ventures/SuperStock

FIGURE 10.17 A model of cytochrome C

TABLE 10.4 Differences in amino acids in cytochrome C between humans and other species

SPECIES COMPARED WITH HUMANS	NUMBER OF DIFFERENCES FROM HUMAN CYTOCHROME C
Chimpanzee	0
Gorilla	0
Rhesus monkey	1
Patas monkey	1
Rabbit	9
Cow	10
Pigeon	12
Bullfrog	18
Tuna	21
Fruit fly	24
Yeast	44

Activity 10.4
Investigating amino acid sequencing

Other protein sequences have been examined and yielded similar results. The alpha and beta chains of the blood protein haemoglobin are identical in humans and chimpanzees, but the same protein sequences in gorillas differ by one amino acid. When the same chains are examined in gibbons, there are three amino acid differences. A comparison of the delta chain indicates that humans differ from chimpanzees and gorillas by one amino acid, and from gibbons by two. Such protein studies provide more support for the evolutionary relationships between primates that have already been established by DNA comparisons.

Bioinformatics

Bioinformatics has become an important part of many areas of human biological science and is particularly useful in providing evidence for evolutionary relationships. It is a multidisciplinary field that combines all areas of biological science with computer science, engineering, statistics and applied mathematics to help understand biological processes. However, in practical terms, **bioinformatics** is the use of computers to describe the *molecular* components of living things. It uses biochemical analysis to gain information about DNA and proteins, and computer software to store and analyse it. Bioinformatics has been particularly useful in assisting evolutionary biologists to trace the evolution of a large number of organisms by measuring changes in their DNA, rather than through traditional techniques of physical taxonomy or physiological observations. The more similar the genes of two species, the closer their evolutionary relationship.

Comparison of molecules
This website presents evidence to show that comparisons of molecules provide support for evolution.

More recent developments have enabled researchers to compare entire genomes. In doing so, the genes and other biological features in a DNA sequence need to be identified, in a process termed **annotation**. This process needs to be computerised, as most genomes are far too large to be annotated by hand. Annotation is made possible by the fact that genes have recognisable start and stop codons (see Chapter 9 of *Human Perspectives ATAR Units 1 & 2*).

Questions 10.3

RECALL KNOWLEDGE

1. State the relationship between DNA, RNA, amino acids and proteins.
2. How many different amino acids make up proteins?
3. When comparing amino acid sequences, scientists use a single letter rather than the three letters that are usually used to identify them. Why do scientists do this?
4. Ubiquitous proteins are important when comparing proteins.
   a. Define 'ubiquitous protein'.
   b. Give an example of a ubiquitous protein.
5. What is bioinformatics?
6. What is annotation, and why is it part of bioinformatics?

APPLY KNOWLEDGE

7. Evolution results from changes in DNA. Given this fact, explain why a comparison of the sequence of amino acids in a particular protein can provide evidence for evolution.
8. Table 10.5 sets out an amino acid sequence from alpha haemoglobin of five different species of animals. Compare each of the amino acid sequences to the one from humans.
   a. Which species' sequence is the most similar?
   b. Which species' sequence is the most different?
   c. Does this correlate with our current understanding of evolution and common ancestors?

TABLE 10.5 The amino acid sequence for alpha haemoglobin of different species

SPECIES	AMINO ACID SEQUENCE FROM ALPHA HAEMOGLOBIN
Human	VLSPADKTNVKAAWGKVGAHAGEYGAEALERMFLSFPTTK TYFPHFDLSHGSAQVKGHGKKVADALTNAVAHVDDMPNAL SALSDLHAHKLRVDPVNFKLLSHCLLVTLAAHLPAEFTPAVH ASLDKFLASVSTVLTSKYR
Whale	VLSPTDKSNVKATWAKIGNHGAEYGAEALERMFMNFPSTKT YFPHFDLGHDSAQVKGHGKKVADALTKAVGHMDNLLDALS DLSDLHAHKLRVDPANFKLLSHCLLVTLALHLPAEFTPSVHA SLDKFLASVSTVLTSKYR
Macaw	VLSGSDKTNVKGIFSKIGGQAEDYGAEALERMFATFPQTKTY FPHFDVSPGSAQVKAHGKKVAAALVEAANHIDDIATALSKLS DLHAQKLRVDPVNFKLLGQCFLVVVAIHNPSALTPEVHASLD KFLCAVGNVLTAKYR
Baboon	VLSPDDKKHVKAAWGKVGEHAGEYGAEALERMFLSFPTTKT YFPHFDLSHGSDQVNKHGKKVADALTLAVGHVDDMPQALSK LSDLHAHKLRVDPVNFKLLSHCLLVTLAAHLPAEFTPAVHASL DKFLASVSTVLTSKYR
Frog	HLTADDKKHIKAIWPSVAAHGDKYGGEALHRMFMCAPKTKT YFPDFDFSEHSKHILAHGKKVSDALNEACNHLDNIAGCLSKLS DLHAYDLRVDPGNFPLLAHQILVVVAIHFPKQFDPATHKALD KFLVSVSNVLTSKYR

CHAPTER 10 ACTIVITIES

Electrophoresis simulation

ACTIVITY 10.1 Investigating electrophoresis simulation

Go to the weblink and work through the electrophoresis simulation. As you go, or after you have finished, answer the following questions.

1. What ingredients are used to make the gel?
2. Describe how the gel is made using the ingredients that you have listed.
3. DNA samples are placed in wells in the gel. Explain how the wells are made.
4. What is the purpose of the DNA size standard?
5. What electrical charge does a DNA molecule have?
6. Which electrical charge is applied to the well end of the gel?
7. Is it possible to tell whether an electric current is running through the gel?
8. What makes the DNA migrate through the gel?
9. Describe the technique that is used to make the DNA visible in the gel.
10. Why do shorter DNA strands move further through the gel than longer strands?

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ACTIVITY 10.2 Investigating the effect of restriction digestion enzymes on lambda DNA

Restriction digestion is the process of cutting DNA molecules into smaller pieces with special enzymes called restriction endonucleases or restriction enzymes. These special enzymes recognise specific sequences in the DNA molecule (e.g. EcoRI GAATTC) wherever that sequence occurs in the DNA.

Aim

To use restriction enzymes to cut DNA into respective fragments
To analyse your restriction digestion using a gel electrophoresis apparatus
Time requirement: 55 minutes

You will need

Restriction digestion materials

Lambda DNA 1 µg (8 uL); restriction digestion buffer (20 µL); EcoRI enzyme (1 µL); HindIII enzyme (1 µL); BamHI enzyme (1 µL); sterile nuclease-free water (200 µL); 4 sterile microtubes 0.5 mL; variable micropipette (2–20 µL); variable micropipette (0.5–10 µL); sterile pipette tips; water bath; microcentrifuge (optional); clock or stopwatch; disposable gloves

Electrophoresis materials

TBE buffer (25 µL); agarose; MIDORI Green safe stain (for pre-staining technique); loading dye 6x (50 µL); variable micropipette (2–20 µL); electrophoresis chamber (blueGel); power supply 100 V (if using an alternative to blueGel); blue light transilluminator (optional)

Note: The above measurements are based on using a blueGel electrophoresis apparatus. If an alternative electrophoreses chamber is being used, increase TBE quantities based on chamber size.

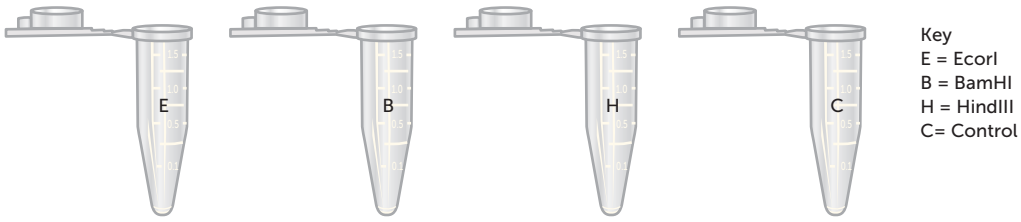
Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
TBE buffer can cause skin irritation.	Wear appropriate personal protective equipment at all times, including eye protection and gloves. Wash skin immediately if contact does occur.
Disposable gloves may pose an allergy risk.	Use a type of glove that removes allergy risk and is suitable for the chemicals being used.

What to do

Restriction digestion

1 Collect four 500 µL (0.5 mL) microtubes and label them as follows:



2 Using a micropipette, add 42 µL (microlitres) of nuclease-free water to each of the microtubes.
3 Add 2 µL of Lambda DNA to each of the microtubes.
4 Using a fresh micropipette tip, add 5 µL of restriction digestion buffer to each of the microtubes.
5 Using a fresh micropipette tip for each sample, add 1 µL of the EcoRI enzyme to 'E', 1 µL of the BamHI enzyme to 'B', 1 µL of the HindIII enzyme to 'H' and 1 µL of nuclease-free water to 'C'.
6 Mix the samples thoroughly by pipetting up and down a few times using the larger micropipette with fresh tips for each sample. Continue until the samples have an even consistency. To collect the liquid at the base of the tubes, spin with a microcentrifuge, or tap the tubes on a bench.
7 Place the microtubes in a 37°C water bath for 10 minutes.

Analysing your digestion using gel electrophoresis

1 Collect the four tubes from the water bath and add 10 µL of loading dye to each sample.
2 Mix samples thoroughly by pipetting up and down a few times using the larger micropipette with fresh tips for each sample, until the solutions look consistent throughout. To collect the liquid at the base of the tubes, spin with a microcentrifuge. Your samples are now ready to be loaded into the gel.
3 Carefully remove the combs from the gel. Place the prepared 0.8% agarose gel into the gel electrophoresis chamber, ensuring that the wells are at the top or negative electrode section of the chamber.
4 Pour TBE buffer into your gel electrophoresis chamber, ensuring that you completely cover the surface of the gel.
5 Using a fresh pipette tip for each sample, load 10 µL of each of your restriction digest samples into the wells located near the negative electrode and note the specific lanes in which the different samples were loaded.
6 Once complete, carefully place the lid on the gel chamber and press the on button and let it run for 30 minutes. Turn on the built-in blue light to visualise DNA band separation if using a blueGel™ electrophoresis chamber.

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Note: If using a gel electrophoresis chamber that requires an external power supply, carefully plug the positive and negative electrodes into the gel box without dislodging the gel. The negative end should be connected to the end closest to the DNA samples. Plug in the power source (set at 100 V), turn it on and let the gel run.

7 After 30 minutes, turn the power supply off and observe the DNA fragments either by turning on blue light or transferring to a blue light transilluminator. Record your results.

Note: If you are using the post-stain method, DNA will not be visible until the gel has been soaked in methylene blue or equivalent for up to 24 hours.

Studying your results

1 How many cuts did each restriction enzyme make?
2 Measure the distance in mm and copy and complete the table below.
3 Graph your results for HindIII digest to determine the sizes of the EcoRI digest and/or BamHI digest. (Try graphing the log – base pairs – vs distance.)
4 Do those fragments add up to the size of lambda DNA? If not, provide possible explanation(s) as to why not.

Analysis of restriction digests of DNA

HindIII		EcoRI			BamHI		
DISTANCE (MM)	SIZE (BP)	DISTANCE (MM)	CALCULATED BP	SIZE (BP)	DISTANCE (MM)	CALCULATED BP	SIZE (BP)
	23 130			21 226			16 841
	9 416						
	6 557						
	4 361						
	2 322						
	2 027						

Discussion

1 Why was 1 µL of nuclease-free water to be added to microtube labelled 'C'?
2 Why do we incubate the restriction digests at 37°C?
3 What is the purpose of the dye?
4 What would occur if the gel electrophoresis chamber were filled with distilled water instead of TBE buffer?
5 Explain why DNA samples must be loaded at the negative end of a gel electrophoresis chamber.
6 What would occur if the electrodes in the electrophoresis chamber were reversed?

Taking it further

Investigate real-world examples of where restriction enzymes are used and how they assist in providing evidence for evolution.

ACTIVITY 10.3 Investigating biotechnological techniques

Throughout this course in human biology, you have had the opportunity to do many activities that have enabled you to inquire scientifically. This activity will allow you to apply some of those skills to investigate a particular biotechnological technique and create your own model of the process.

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What to do

Working with a partner, or as part of a small group, select either the polymerase chain reaction or DNA sequencing for further investigation. Both techniques have a number of stages that provide logical steps to allow models to be created.

1. Use a variety of references to establish the exact sequence of steps in the technique being investigated. Collate the information about your sources in a bibliography.
2. Draw a diagram to clearly illustrate all the steps in the process.
3. A scientific model is a simplified representation of an idea or a process. Using different shapes cut out of cardboard to represent the different parts of the process, build a simple model to demonstrate how the technique you are investigating takes place. Depending on the technique being investigated, your shapes may represent various nucleotides or segments of the DNA molecule. There will be many different ways of presenting the model, so do not be surprised if yours is quite different from others' models.
4. Present and explain your model to the other members of the class, or make a video of your model and add an explanation by a voice-over or annotations.

ACTIVITY 10.4 Investigating amino acid sequencing

Haemoglobin is the protein that carries oxygen in the blood. It is found in all mammals and has the same function in each species. You would expect that it would be composed of the same sequences of amino acids. However, this is not the case. The particular protein chain we will study in this activity is composed of 146 amino acids. The numbers in the following table indicate the position of some of the amino acids in that sequence and the letters are abbreviations for the amino acids (see Table 10.3). Six different mammalian species are shown with the amino acids that are present at positions 87 to 116 in the chain.

What to do

1. Examine the table on the following page and count the number of differences in the amino acid sequences for the following pairings of species:
   - human and chimpanzee
   - human and gorilla
   - chimpanzee and gorilla
   - human and rhesus monkey
   - chimpanzee and rhesus monkey
   - gorilla and rhesus monkey
   - human and horse
   - human and kangaroo.
2. Record your data in a table.
3. Using only the data from this section of the haemoglobin molecule, rank the species in order from the one closest to humans to the one most distant.

Studying your data

1. Based on this segment of the haemoglobin molecule, which species of mammal appears to be the most closely related to humans?
2. Which animal appears to be the least closely related to humans?
3. Which of the other pairs of species show close relationships?
4. These sequences of amino acids are generally very similar but not identical. If these species were all descended from a common ancestor, how would the changes in the sequences of the different species have come about?
5. Do you think the differences in the amino acid sequences between the species would affect the function of haemoglobin?

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Amino acid sequences in the haemoglobin of six mammalian species

AMINO ACID IN HAEMOGLOBIN	SPECIES					
	HUMAN	CHIMPANZEE	GORILLA	RHESUS	HORSE	KANGAROO
87	T	T	T	Q	A	K
88	L	L	L	L	L	L
89	S	S	S	S	S	S
90	E	E	E	E	E	E
91	L	L	L	L	L	L
92	H	H	H	H	H	H
93	C	C	C	C	C	C
94	D	D	D	D	D	D
95	K	K	K	K	K	K
96	L	L	L	L	L	L
97	H	H	H	H	H	H
98	V	V	V	V	V	V
99	D	D	D	D	D	D
100	P	P	P	P	P	P
101	E	E	E	E	E	E
102	N	N	N	N	N	N
103	F	F	F	F	F	F
104	R	R	K	K	R	K
105	L	L	L	L	L	L
106	L	L	L	L	L	L
107	Q	Q	Q	Q	Q	Q
108	N	N	N	N	N	N
109	V	V	V	V	V	I
110	L	L	L	L	L	I
111	V	V	V	V	A	V
112	C	C	C	C	L	I
113	V	V	V	V	V	C
114	L	L	L	L	V	L
115	A	A	A	A	A	A
116	H	H	H	H	R	E

In summary

Using the information from this sequence of amino acids in haemoglobin, describe the evolutionary relationships between the species in terms of the evolution of humans.

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CHAPTER 10 SUMMARY

- Polymerase chain reaction (PCR) is used to make many copies of a section of DNA.
- PCR involves denaturing DNA to separate the strands, annealing primers to the DNA and then extension of the strands.
- Primers are short sections of DNA with a base sequence that is complementary to sections on either side of the DNA that need to be copied.
- DNA polymerase is used to add nucleotides to the growing strand. As the enzyme used needs to be able to withstand the high temperatures during denaturing, Taq polymerase from a heat-loving bacterium is used.
- Gel electrophoresis is used to separate DNA strands based on their length.
- DNA has a negative charge. Therefore, when an electric current is applied to the gel, the DNA will move away from the negative terminal. Smaller lengths of DNA will move more quickly than longer lengths. This will produce a DNA profile.
- Restriction enzymes are used prior to gel electrophoresis to cut the DNA at specific sites.
- DNA ladders are produced by running a sample with known DNA lengths through gel electrophoresis. Results from an unknown sample can be compared to the ladder, giving information about the length of the DNA strands.
- Ethidium bromide, methylene blue or DNA probes are used to visualise the DNA after gel electrophoresis.
- The sequence of nucleotides (or the bases on the nucleotides) can be determined using Sanger's method, which uses dideoxyribonucleotides to stop the extension of the DNA strand. The strands are then separated using gel electrophoresis. Knowing which base was on the dideoxyribonucleotide for each strand length allows the base sequence to be determined.
- Ethical factors such as autonomy, confidentiality, equity and privacy need to be considered when gaining and using genetic information.
- Comparative genomics is the comparison of the DNA of different species. Species with a recent common ancestor will have more similarities than those with a more distant common ancestor.
- Endogenous retroviruses (ERV) are viral sequences that become part of the organism's DNA in a gamete. Comparison of the amount and location of ERV of different species can provide evidence for common ancestors.
- Mitochondrial DNA (mtDNA) is circular DNA found in the mitochondria and is passed down from the mother. The degree of mutation can be used to determine the closeness of the relationship between species.
- The amino acid sequence of ubiquitous proteins such as cytochrome C can be compared and used to infer how recently the common ancestor of two species existed.
- Bioinformatics uses computers to describe the molecular components of a living organism. For example, it can compare the whole genome of species.

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CHAPTER 10 GLOSSARY

Annotation Identification of genes in a DNA sequence

Bioinformatics The use of computers to describe the molecular components of living things

Comparative genomics The comparison of genome sequences of different species

Cytochrome C An iron-containing protein that can alternate between a reduced form and an oxidised form; important in the electron transport system in cellular respiration

Dideoxyribonucleotide A modified deoxyribonucleotide that lacks a hydroxyl group of the sugar component

DNA fingerprint A technique that uses the banding patterns of DNA fragments as a means of identification; a DNA fingerprint is unique to a particular individual; also called DNA profile

DNA profile *see* DNA fingerprint

DNA sequencing The determination of the precise order of nucleotides in a sample of DNA

Endogenous retrovirus (ERV) A retrovirus that has become part of an organism's genome and exists in every cell of the body

Gel electrophoresis A process used to separate charged molecules based on their size by pushing them through a gel

Genome The complete set of genetic material in a cell; an organism's complete set of DNA

Micropipette A fine pipette used to measure and transfer very small volumes of liquid

Mitochondria Structures in the cytoplasm of a cell in which the aerobic stage of respiration occurs; singular: mitochondrion

Mitochondrial DNA (mtDNA) DNA found in the mitochondria of the cells, rather than in the nucleus

Polymerase chain reaction (PCR) A technique used in molecular biology for producing multiple copies of DNA from a sample; used in DNA fingerprinting and in identifying diseases

Primer A strand of DNA or RNA that serves as a starting point for DNA replication

Restriction enzyme An enzyme that cuts strands of DNA at a specific sequence of nucleotides

Reverse transcription A process where the base sequence in RNA is copied during the synthesis of DNA

Thermocycling A process of repeated heating and cooling

Ubiquitous protein One of a group of proteins that appears to be in all species, from bacteria to humans; the small protein called ubiquitin was so named because it is present in all types of cells

Well An indentation in the gel used for gel electrophoresis

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CHAPTER 10 REVIEW QUESTIONS

Recall

1 a What is DNA sequencing and what is it used for?
b Briefly outline the steps in building a DNA sequence.

2 a What is a 'DNA profile'?
b List two practical applications of DNA profiling.

3 a Outline the steps in the polymerase chain reaction.
b Giving an example, explain what the term 'heat stable DNA polymerase' means.
c What are some of the practical applications of the polymerase chain reaction?

4 a Define 'endogenous retroviruses'.
b How do retroviruses become endogenous?
c What is the value of endogenous retroviruses in a study of evolution?

5 a Define 'mitochondrial DNA (mtDNA)'.
b Describe how mtDNA has been used to provide evidence for evolutionary relationships between species.
c Give an example of where mtDNA has provided information about such a relationship.

6 Describe how the sequence of amino acids in proteins can be used to determine the degree of similarity between species.

7 a Define 'ubiquitous proteins'.
b Why has cytochrome C been so valuable in providing evidence for evolution? Give examples of species that contain cytochrome C.
c Besides cytochrome C, what other proteins have been used to provide evidence about relationships between species?

8 a How has bioinformatics assisted biologists in refining evolutionary relationships?
b What role has comparative genomics played in the study of evolutionary changes among organisms?

9 List the key areas that need to be ethically considered when deciding whether to gain or use genetic information.

Explain

10 One of the most frequently used ways to sequence DNA is to take advantage of the way it replicates. Explain how, if the sequence of bases on one side of a fragment of DNA is known, the sequence on the other side is known as well.

11 The polymerase chain reaction is a method of amplifying a small amount of DNA into a much larger amount. Explain the advantages of being able to do this.

12 Using an example, explain how the study of DNA in different species has added to the evidence for evolution.

13 Describe how each of the following has facilitated DNA sequencing:
a polymerase chain reaction
b gel electrophoresis
c bacterial enzymes.

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Apply

14 When ancestral species evolve into two or more separate species, those new species would exhibit considerable similarity in their DNA. What causes the DNA to change over time? How has the information from DNA been used by scientists to speculate on the relationships between species?

15 Modern technology has provided the means to compare DNA and protein sequences. How has this changed the traditional way of looking at the relationships between humans and apes?

16 Explain why mtDNA is only of use when looking at the relationships within a species or between closely related species.

17 Explain why scientists select ubiquitous proteins for their biochemical research on the relationships between species.

18 Refer to Table 10.4, which indicates the degree of difference in the amino acids in cytochrome C between humans and some other species. Using this information, construct a family tree to illustrate a possible relationship between those species.

19 Why would scientists use a comparative study of haemoglobin in different species in a search for data to support their theories of primate evolution?

20 Imagine that you and your sister are identical twins. You have had genetic testing, and now know that you have a genetic predisposition to breast cancer. Discuss whether or not it is ethical to tell your twin about the testing results.

Extend

21 Mitochondrial Eve is a name that has been given to the woman who, when traced through the female line, is the most recent common ancestor for all living humans. The mitochondrial DNA in all humans alive today is derived from her.

- **a** How is the matrilineal line traced back to Mitochondrial Eve?
- **b** How long ago is Mitochondrial Eve believed to have lived?
- **c** In what part of the world did she live?
- **d** Does the fact that the mitochondrial DNA of all humans is derived from Mitochondrial Eve mean that she was the only human female alive at the time?
- **e** How is it possible that one woman could be the matrilineal ancestor of us all?

22 Haemoglobin and cytochrome C have been used to give support to the theory of evolution through natural selection. Scientists have similarly compared the biochemistry of universal blood proteins.

- **a** Have such studies revealed evidence for the relationships between different species?
- **b** Does such evidence imply that some species share a more recent common ancestor than other species?

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11 OTHER EVIDENCE OF EVOLUTION

UNIT 4 CONTENT

SCIENCE INQUIRY SKILLS

- conduct investigations, including the use of virtual or real biotechnological techniques of polymerase chain reaction (PCR), gel electrophoresis for deoxyribonucleic acid (DNA) sequencing, and techniques for relative and absolute dating, safely, competently and methodically for valid and reliable collection of data
- select, use and/or construct appropriate representations, including phylogenetic trees, to communicate conceptual understanding, solve problems and make predictions

SCIENCE AS A HUMAN ENDEAVOUR

- developments in the fields of comparative genomics, comparative biochemistry and bioinformatics have enabled identification of further evidence for evolutionary relationships, which help refine existing models and theories

SCIENCE UNDERSTANDING

Evidence for evolution

- comparative studies of DNA (genomic and mitochondrial), proteins and anatomy provide additional evidence for evolution; genomic information enables the construction of phylogenetic trees showing evolutionary relationships between groups
- the fossil record is incomplete and cannot represent the entire biodiversity of a time or a location due to many factors that affect fossil formation, the persistence of fossils and accessibility to fossilised remains
- sequencing a fossil record requires a combination of relative and absolute dating techniques to locate fossils onto the geological time line
- both relative dating techniques, including stratigraphy and index fossils, and absolute dating techniques, including radiocarbon dating and potassium–argon dating, have limitations of application

Source: School Curriculum and Standards Authority, Government of Western Australia

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The living things that exist on Earth today have all evolved from simpler forms that existed in the past. In Chapter 10 you learnt about how we can use DNA to show the gradual changes in an organism over time. However, our understanding of evolution started before we were able to analyse and compare DNA. One of the crucial pieces of evidence for evolution is the record of those changes left to us in the form of fossils.

Alamy Stock Photo/Science Photo Library

FIGURE 11.1 The fossil hominin skeleton known as 'Lucy'

11.1 FOSSILS

A **fossil** is any preserved trace left by an organism that lived long ago. Some fossils are bones, shells or teeth. However, fossils do not have to be a part of an organism; they also include footprints, burrows, faeces, or impressions of all or part of an animal or a plant. In the case of human ancestors, fossil remains are usually bones, teeth or sometimes footprints. Figure 11.1 shows the fossil remains of an ancestor of modern humans, *Australopithecus afarensis*, found in the Hadar region of Ethiopia. Such fossil remains are extremely important as they allow scientists to determine exactly what extinct species were like. Other material associated with the bones, such as the rock in which they were found and fossils

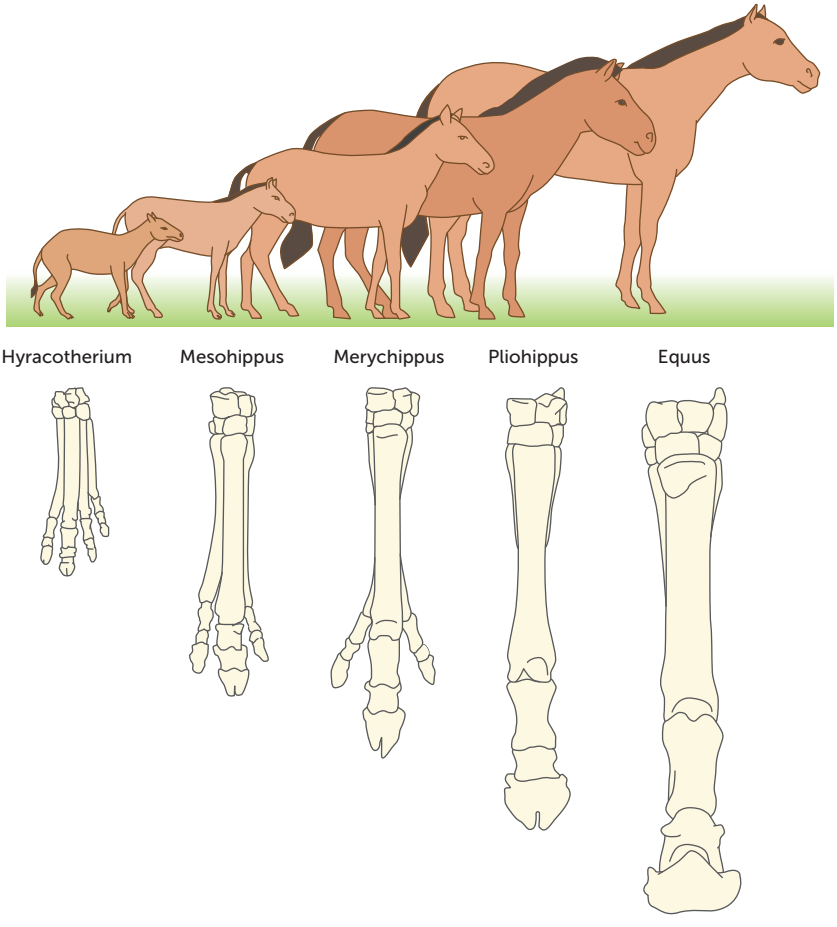

FIGURE 11.2 The evolution of the horse, showing how the leg bones appear to have changed as the animal increased in size

Horse evolution
This website provides more information about the evolution of the horse. Click on 'Gallery of horse fossils'.

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of other plants and animals, allows the scientist to develop a picture of life in the past – what the organisms ate, what other organisms existed at that time and, sometimes, even what the climate was like.

There are many cases where the fossil record has allowed scientists to build up a sequence of the evolution of a particular plant or animal. One of the best known is the evolution of the horse, which can be traced through fossil remains from a small creature not much bigger than a small dog to the horses that we know today.

Fossil formation

When we consider the billions of organisms that have lived on Earth, the chance that a plant or animal will be fossilised is very small. Normally, dead organisms are decayed by micro-organisms and no trace of their existence is left. Parts of organisms may become fossilised when buried by drifting sand, mud deposited by rivers, volcanic ash or, in the case of some of the more recent human ancestors, other members of the species. If buried rapidly, conditions may not be suitable for the activity of decay organisms (decomposers), and decomposition may be slowed or prevented.

Effect of soil type on fossilisation

The nature of the soil is very important for the fossilisation of bone. In wet, acidic soils the minerals in the bone are dissolved and no fossilisation occurs. However, if such soil contains no oxygen, as in the case of peat, complete preservation of the soft tissues and bones of the animal may occur. Bones buried in alkaline soils produce the best fossils because the minerals in the bones are not dissolved. New minerals, often lime or iron oxide, are deposited in the pores of the bone, replacing the organic matter that makes up about 35% by weight of the bone. The bone becomes petrified (turned into rock), but the details of structure are still preserved.

Location of fossils

Fossils of human ancestors are often found at the edges of ancient lakes and river systems, in caves or in volcanically active areas. This is because the organism can be buried rapidly, preventing decomposition. Lakes and rivers build up sediments when flooding occurs or when the water flow slows rapidly. Many caves are in limestone, which consists of calcium carbonate. This chemical may be deposited around dead organisms, or the cave roof or walls may collapse, covering the bodies of animals. It is unusual for animals to be preserved near volcanic eruptions because heat from the volcanic material destroys the organism, but in East Africa ash falls have preserved fossils of many human ancestors.

Alamy Stock Photo/UPI

FIGURE 11.3
Fossilised remains of *Homo sapiens* were found in a cave called Jebel Irhoud in Morocco

Discovery of fossils

Fossils are sometimes found by chance at the surface of the ground where they may have been uncovered by erosion, but more often the discovery of fossils is the result of slow and painstaking excavation of likely sites. Surface discoveries such as fossil fragments, or evidence of human occupation such as that found in many caves, are indications of places where excavations may prove fruitful. Scientists refer to an excavation as a 'dig'.

The area to be investigated is first surveyed and marked out in sections. Small hand tools are used to remove the soil gently so as not to damage any of the material. The soil removed is usually sieved so that even very small fragments are not overlooked. In the case of fossils of human ancestors, artefacts are often found in association with the fossils. **Artefacts** are objects that have been deliberately made by humans. They include items such as stone tools, beads, carvings, charcoal from cooking fires, and cave paintings.

Alamy Stock Photo/Ken Gillespie Photography

FIGURE 11.4 Archaeologists work with small hand tools to carefully uncover fossils and other artefacts at a site in Canada

Photographs are taken at every stage of a dig so that detailed studies of the positions of uncovered material can be carried out later. Each item is carefully labelled and catalogued for the prolonged study that follows the excavation of the site. In the laboratory, fossil bones are carefully scraped clean, broken parts are pieced together, measurements are taken, and plaster casts or latex moulds may be made.

Fossils show human migration
This website explains how recently discovered fossils show that humans migrated from Africa 180 000 years ago.

Science Photo Library/Pasquale Sorrentino

FIGURE 11.5 Cleaning prehistoric animal bones. These bones are from the jaw of a rhinoceros dating back 600 000 years

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Key concept

Fossils are any preserved trace left by an organism that lived long ago. They form when organisms, artefacts or imprints are covered rapidly and left undisturbed, and conditions such as alkaline soils, a lack of oxygen and an absence of decay organisms prevent decomposition.

Questions 11.1

RECALL KNOWLEDGE

1 What is a fossil?
2 List the conditions needed for fossils to form.
3 Explain why fossils are often found near lakes and rivers.
4 Why are hand tools used to dig at excavation sites rather than bigger earthmoving equipment?
5 Describe artefacts, including at least one example.

APPLY KNOWLEDGE

6 Why are fossil remains of organisms usually bones or teeth, and not the skin, muscle or organs?
7 Fossils of soft tissue are most likely to be found in acidic soil, while the best fossils of bones are found in alkaline soils. Explain why this occurs, and include any other conditions that must be present for each of these fossils to form.

11.2 DATING FOSSILS

One of the major tasks following the excavation of fossils or artefacts is to determine the age of the material. This is known as **dating**. Knowledge of the age is crucial in finding out the sequence of changes that have resulted in present-day humans. Various methods of dating fossils and material associated with human ancestors have been devised. Dating can provide:

- **absolute dates** – the actual age of the specimen in years
- **relative dates** – a comparison of fossils to tell us whether one sample is older or younger than another.

Modern technology has enabled accurate estimates to be made of the absolute age of many samples. However, when that is not possible, knowing whether one fossil is older or younger than another is very important.

The age (or date) of a fossil or artefact is usually given in years before the present time. For example, a fossil may be said to date from 45 000 years BP, which is another way of saying it is 45 000 years old. BP stands for 'before present'.

Absolute dating

Potassium–argon dating

Potassium is one of the most abundant elements in Earth's crust, and is therefore found in some rocks. The **potassium–argon dating** technique is based on the decay of radioactive potassium to form calcium and argon. Potassium (chemical symbol K) is a mixture of three different isotopes. **Isotopes** are atoms of the same element with different numbers of neutrons. Isotopes of potassium all have 19 protons, but atoms of potassium-39 have 20 neutrons, atoms of potassium-40 have 21 neutrons, and atoms of potassium-41 have 22 neutrons. Potassium-40 is a radioactive isotope and decays to form calcium-40 and argon-40. Such decay takes place at an extremely slow, but constant rate, and so determining the amounts of potassium-40 and argon-40 in a rock sample enables the age of the rock to be calculated. As the rock ages, the proportion of potassium-40 decreases while that of argon-40 increases.

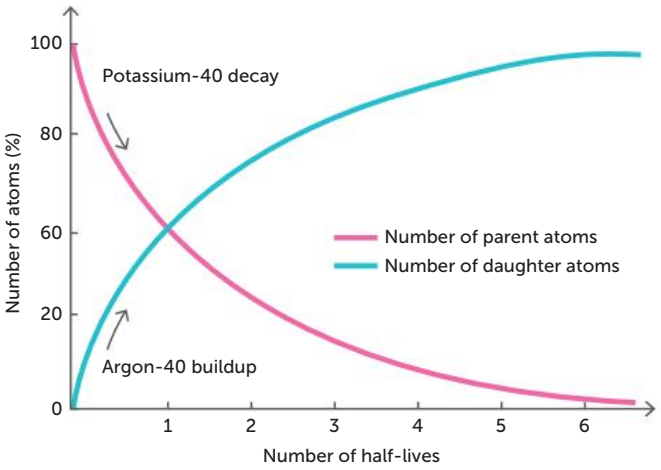

FIGURE 11.6
The levels of potassium-40 and argon-40 as a rock ages

Potassium–argon dating
This website provides more information about potassium–argon dating and some animated sequences on dating.

Potassium–argon dating has limited usefulness: not all rock types are suitable for this method of dating and it can only date rocks older than 100 000 to 200 000 years. The **half-life** of potassium-40 is 1250 billion years (1.25×10^9 years). This means that it takes 1250 billion years for half of the potassium-40 to decay. Therefore, after 100 000 years, only 0.0053% of the potassium-40 in a rock would have decayed to argon-40. Such a small amount pushes the limits of detection devices currently in use. Therefore, potassium–argon dating is most useful on samples that are older than 200 000 years.

To determine the age of a fossil using this method, some suitable rock of the same age as the fossil must be available. The age of the rock is determined using potassium–argon dating, and hence the age of the fossil is inferred. This situation can occur when rocks produced in volcanic eruptions bury bones.

Carbon-14 dating

The **carbon-14** or **radiocarbon dating** method is based on the decay of the radioactive isotope of carbon, carbon-14, to nitrogen. Carbon-14 is produced in the upper atmosphere by the action of cosmic radiation on nitrogen at about the same rate at which it decays. In the atmosphere there is a ratio of one carbon-14 atom to every million million (10^{12}) atoms of the stable isotope carbon-12.

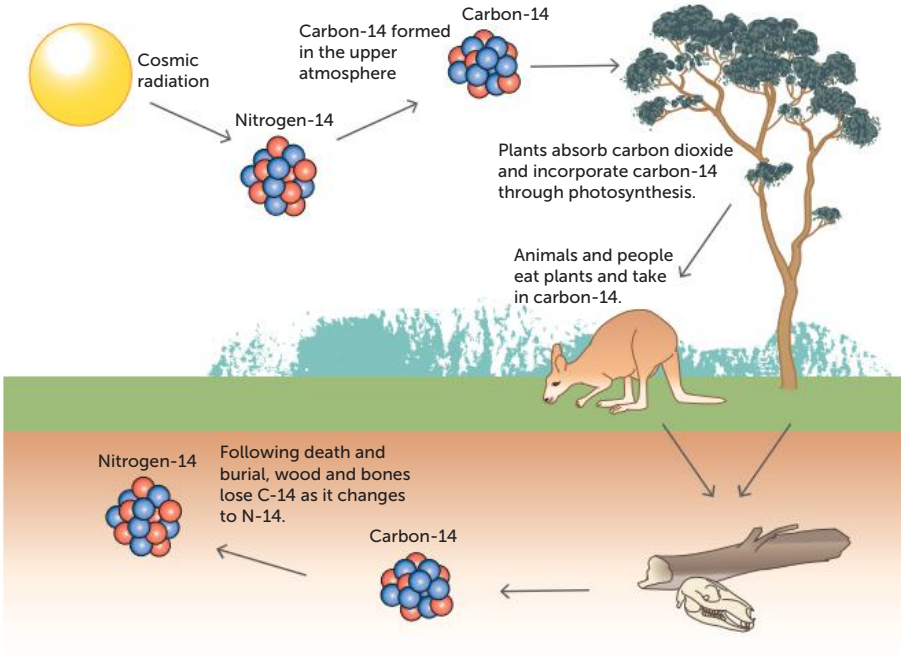

FIGURE 11.7
Summary showing how carbon-14 is formed, enters living things and decays

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When green plants use atmospheric carbon dioxide in photosynthesis, one atom in every million million of the carbon atoms incorporated in the plant tissues is carbon-14. Should an animal eat the plant, the carbon-14 then becomes a part of the animal's tissues. With death, an organism's intake of carbon-14 ceases, but the carbon-14 already in the tissues of the organism continues to decay at a fixed rate. By measuring the amount of radiation liberated by a sample, the ratio of carbon-14 to carbon-12 can be estimated, and from this the age of the sample can be calculated.

Figure 11.8 shows the rate of decay of carbon-14. The ratio of radioactive carbon in the tissues of a living organism today is one carbon-14 atom to every 10^{12} carbon-12 atoms. This ratio declines to 0.5 in 10^{12} after 5730 years, to 0.25 in 10^{12} after another 5730 years, and so on. In other words, over a period of 5730 $\pm$ 40 years, half of any given quantity of carbon-14 breaks down. Therefore, 5730 $\pm$ 40 years is the half-life of radioactive carbon.

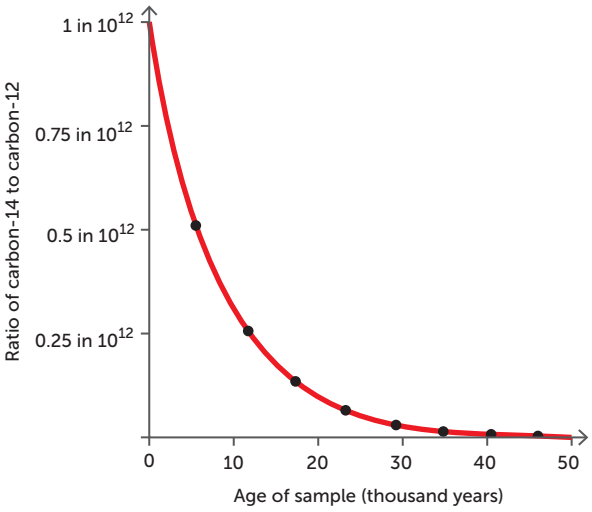

FIGURE 11.8 Rate of decay of carbon-14 to nitrogen. Each dot on the curve represents a period of 5730 years, the half-life of carbon-14

The normal method of radiocarbon dating requires at least three grams of organic material so that the rate of radioactive decay of carbon-14 in the sample can be measured. A more refined technique, known as **accelerator mass spectrometry (AMS) radiocarbon dating**, can be used to date a sample as small as 100 micrograms (0.0001 grams). This technique involves breaking the sample up into its constituent atoms so that the number of atoms of each isotope of carbon can be counted. Using AMS radiocarbon dating, it has become possible to date cave paintings accurately from tiny samples of pigment. It has been found that such pigments often contain organic material such as charcoal, while honey, milk, blood or oil seed may have been used to bind the pigment particles.

After about 70 000 years, the percentage of carbon-14 left is only 0.021%. This is too small to be able to measure accurately; therefore, radiocarbon dating method cannot be used to date back more than about 60 000 years. A further limitation is that the material to be dated must contain **organic compounds**, compounds from living things that contain carbon. Radiocarbon dating is nevertheless of great value in dating fossils of more recent origin and also in dating artefacts, because these are often found in association with charcoal left from cooking fires. By dating the charcoal found in ancient hearths, the approximate age of artefacts can be deduced.

There is another problem with using radiocarbon dating. It was once assumed that the ratio of carbon-14 to carbon-12 in the atmosphere was constant, but it is now known that the amount of carbon-14 in the atmosphere varies. Thus, radiocarbon dates must be treated with a certain degree of caution. Corrections for the fluctuations in the carbon-14 content of the atmosphere are, however, now possible for about the past 9000 years, by reference to other information such as tree-ring dating.

Each method of absolute dating is limited in its application because each depends on the occurrence of a particular set of circumstances before it can be used. Together, however, these and other methods do give the anthropologist a number of ways of determining the actual age of ancient material. New methods are being developed all the time and there is constant improvement in the accuracy of older methods. Table 11.1 shows the approximate time span applicable to the methods described and also those of some more recently developed techniques.

Carbon-14 dating
This website provides an animation of the use of carbon-14 dating.

How carbon-14 dating works
This website provides further explanation of the processes involved in carbon-14 dating.

Activity 11.1
Investigating radioisotope methods of dating

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Key concept

Absolute dating methods, such as potassium–argon dating and carbon-14 dating, allow the age of fossils to be identified. Potassium–argon dating is used to date rocks older than 200 000 years, while carbon-14 dating is used on organic matter less than 60 000 years old.

TABLE 11.1 Useful range of common absolute dating methods

DATING METHOD	MATERIAL USED	USEFUL RANGE (YEARS BP)
Tree growth rings (dendrochronology)	Wood	Up to 9000
Carbon-14	Carbon compounds	Up to 60 000
Protactinium	Sea sediments	Up to 250 000
Uranium–thorium	Sea sediments, coral	Up to 600 000
Potassium–argon	Volcanic deposits	200 000 And earlier
Electron spin resonance	Calcium carbonate (in shells, coral, teeth), also quartz and flint	Up to 100 000, possibly 300 000
Fission tracks	Minerals and glass	100 Years ago to 4550 million
Thermoluminescence	Sediments, lava, ceramics	300 Years ago to 100 000

Relative dating

When it is not possible to determine the actual age of a fossil or artefact, scientists can often determine whether it is older or younger than another sample, or whether it is older or younger than the rock or soil in which it is found. Such relative dating enables a sequence of events to be established.

Stratigraphy

Stratigraphy
This website provides more information about the problems that may arise with stratigraphy.

Stratigraphy is the study of layers, or strata. There are two ways in which stratigraphy can be useful in dating fossil material. The first is the **principle of superposition**, which assumes that in layers of sedimentary rock the layers at the top are younger than those beneath them. Thus, any fossils or other material found in the top layers will be younger than material found lower down. This principle must be applied with care because distortions of Earth's crust do occur, and a sequence of rock layers may be turned upside down. In addition, it is possible for fossils or artefacts to be buried by animals, or perhaps by early humans, some time after the deposition of sediment. In this case, the specimen may be younger than some of the layers above it.

The second use of stratigraphy is in the **correlation of rock strata**, which involves matching layers of rock from different areas. Matching of strata can be done by examining the rock itself, and also by studying the fossils it contains. Rocks that contain the same fossils may be assumed to be of the same age. Certain fossils, called **index fossils**, are of great value in this correlation work because they are widely distributed and were present on Earth for only a limited period of time. This makes the relative dating of strata more precise. Figure 11.11 (on page 304) shows how index fossils may be used to correlate strata from different localities, often hundreds, or even thousands, of kilometres apart.

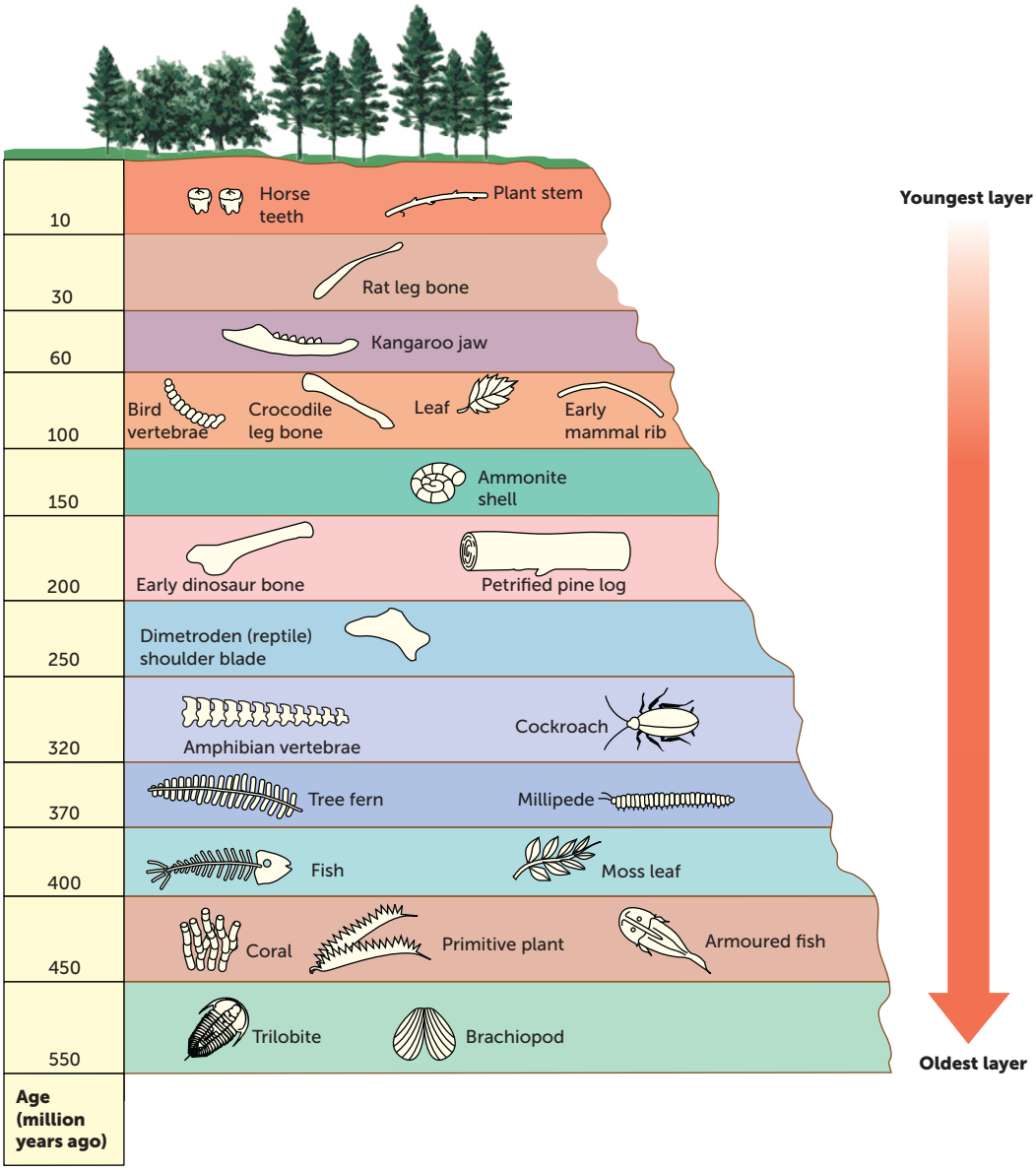

FIGURE 11.9
A section of rock strata containing fossils of different ages. The principle of superposition assumes that the younger strata are towards the top of the sequence

Shutterstock.com/Kristyna Henkeova

FIGURE 11.10
A trilobite fossil. The segmented form of the shell is clearly visible, with the head at bottom left. Trilobites make excellent index fossils as they lived between 500 and 300 million years ago before becoming extinct

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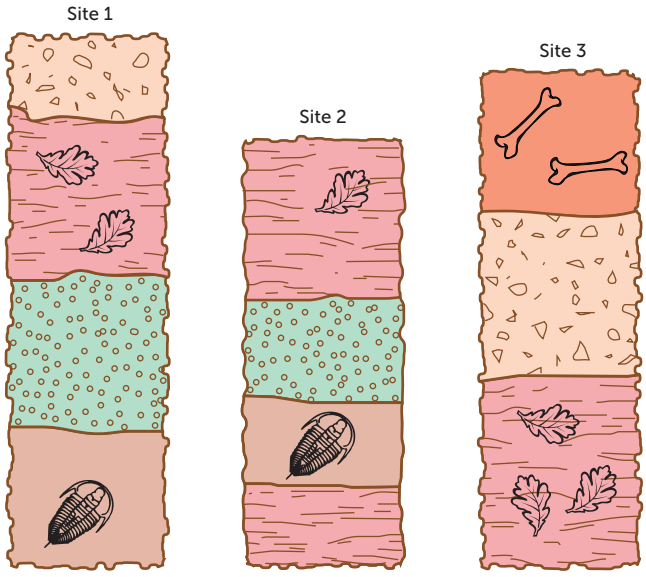


FIGURE 11.11 Three sets of rock strata, some containing fossils exposed in trenches at three different excavation sites. Which of the strata shown is the oldest?

Activity 11.2
Investigating stratigraphy

An analysis of fossilised pollen grains has developed into an important branch of science. Some fossil pollen grains are useful as index fossils but, even if they cannot be used in this way, the presence of preserved pollen grains in a soil or rock sample can enable a botanist to construct a picture of the type and amount of vegetation existing at the time the deposit was laid down. An idea of the climatic conditions prevailing at the time may then be worked out. This data can be used to confirm or refute relative dates arrived at by other methods.

Key concept

Relative dating, such as stratigraphy, allows the age of fossils to be compared. Stratigraphy is based on the principle of superposition, which assumes that layers of sedimentary rock are youngest on the top.

Problems with the fossil record

The fossil record is very incomplete as conditions for fossilisation do not always occur, or occur at irregular periods of time. For fossils to be formed, four conditions are usually required:

- a quick burial of the material
- the presence of hard body parts
- an absence of decay organisms
- a long period of stability – the organism needs to be left undisturbed.

Fossilisation is therefore a chance occurrence and there are many gaps in the fossil record because organisms have not been preserved.

Another reason for gaps in the fossil record is that only a very small proportion of the fossils that do exist have actually been discovered. Some are buried too deep in the ground to be found, or are in inaccessible places. Others may not have been recognised as fossils or may have been inadvertently destroyed by human activity such as agriculture or industry.

Even when fossils are found, dating the fossil material can be problematic. To use carbon dating techniques, material containing carbon must be present, and the material can only be dated back to 60 000 years or so. To date material older than this, the age of the fossil is determined by the sediments in which it is found. For example, the use of potassium–argon dating relies on suitable material, such as volcanic lava, being present.

It is unusual to find a fossil of an entire organism, or the whole skeleton of an organism. This is particularly true of fossils of human ancestors. Often, from just a few fragments of bone, scientists have to reconstruct what the organism may have looked like. Figure 11.12 shows a reconstructed skull of *Homo ergaster*. Found in Kenya, the fragments were dated at around 1.8 million years old, and were pieced together like a jigsaw puzzle. Areas shown in black are parts of the skull where bones are missing. Experts have estimated the shape of the face and upper jaw in order to complete the reconstruction. Such reconstructions are only approximations based on the experience of the scientists involved. Other scientists may disagree with the interpretation and this can lead to considerable controversy. The only resolution to such disagreements is to find more fossils; but as we have seen, even if there are more fossils, the chances of finding them are often slim.

FIGURE 11.12 The reconstructed skull of *Homo ergaster*

Evidence for evolution
This website provides a review of evidence for evolution that has been discussed in this chapter and the previous one.

11.1 Fossil evidence for evolution

Key concept

There are gaps in fossil records because fossils only form in certain conditions, they may be accidentally damaged, they may not yet have been found, or it may not be possible to date them.

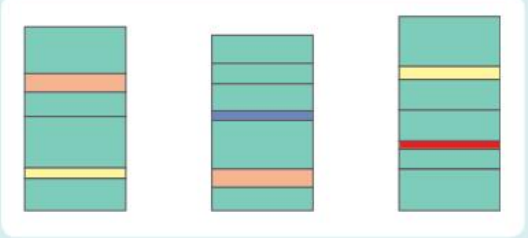
Questions 11.2

RECALL KNOWLEDGE

1. Describe the difference between absolute dating and relative dating.
2. Define 'isotopes' and list three isotopes of potassium.
3. Explain how the amount of potassium-40 can provide information about the age of the sample.
4. What types of samples are able to be dated using potassium–argon dating?
5. What types of samples are able to be dated using carbon-14 dating?
6. Define 'half-life' and state the half-life of carbon-14 and potassium-40.
7. What type of dating could be used to determine the age of a wooden artefact?
8. Explain why the principle of superposition cannot be considered without taking other factors into account.
9. Describe index fossils and explain their relevance to relative dating.
10. List the reasons that there are gaps in fossil records.

APPLY KNOWLEDGE

11. Compare and contrast atoms of potassium-40, calcium-40 and argon-40. Use this to suggest what happens when potassium-40 decays to form calcium-40 and argon-40.
12. Explain why dating using carbon-14 is also called radiocarbon dating.
13. Explain why carbon-14 dating can only be used to determine the age of samples that were once living.
14. The diagram below shows a sample of soil taken from the same area. The colours represent fossils found in particular layers. State the age of the fossils from the youngest to the oldest.



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11.3 COMPARATIVE ANATOMY

Fossil evidence and current observations can provide information about the anatomy of organisms. This allows scientists to compare the structural features of related animals to ascertain the degree of similarity between them. Similarities in structure often suggest that species have a common ancestor. In discussing comparative anatomy, we will focus on three areas:

- **embryology** – comparing the very early stages of the development of organisms
- **homologous structures** – structures that are similar in structure but may be used in different ways
- **vestigial structures** – structures that may once have been important but have lost or changed their function.

Embryology

How fudged embryo illustrations led to drawn-out lies
Learn how *Haeckel's Embryos* misled people about comparative embryology.

Comparative embryology provides evidence for evolutionary change over time by comparing the early stages in the development of organisms. Although it is relatively easy to distinguish between the adults of different species, it is frequently far more difficult to tell the difference between embryos. In vertebrates, comparing the embryonic stages reveals a remarkable similarity between different species at different times.

At one point in their development, the embryos of humans and chickens have slits and arches in their neck. These are similar to the gill slits and arches in fish, but do not develop into gills. However, their similarities indicate a common ancestor.

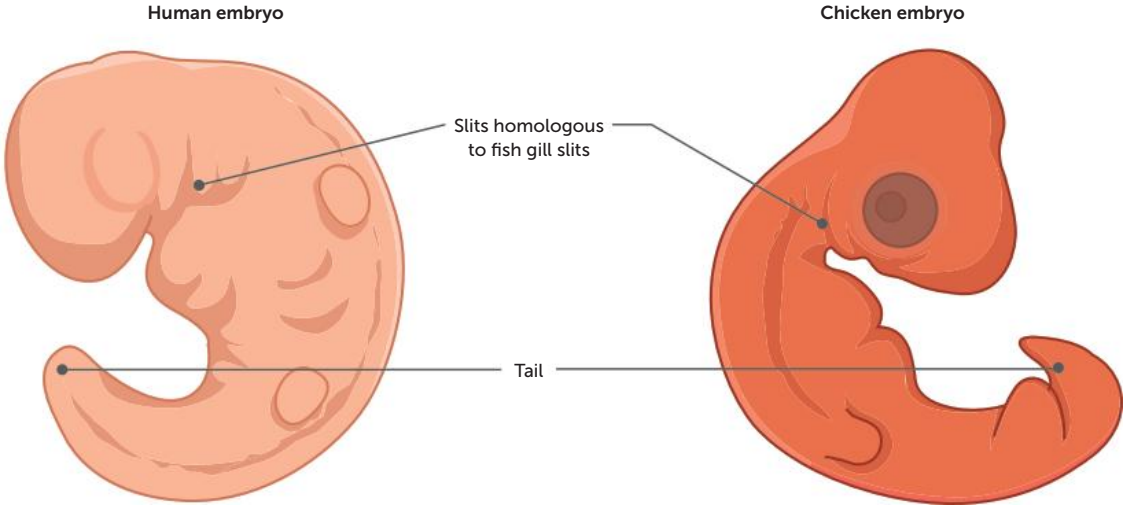

FIGURE 11.13
Comparing the human and chicken embryo at one stage of development

Additional features common to vertebrate embryos at one stage are the presence of a tail, a two-chambered heart and similar brain development. This all adds up to striking evidence for a common ancestry with later evolution along different pathways.

Homologous structures

One of the classic examples used to illustrate similarity in anatomy is the forelimbs of vertebrates. The same bones appear in various forms throughout the vertebrates: the feet of amphibians and reptiles, the wings of bats and birds, the leg of a horse, the flipper of a whale or seal, and the human hand. The degree of similarity between the bones can be seen in Figure 11.14. In every case the bones are arranged in a similar way, even though some have developed different functions. These forelimb bones are described as homologous structures because they possess a similar structure. Organisms possessing homologous structures are likely to have a common ancestor. Therefore, the arrangement of the bones of the forelimb in such a range of vertebrates is convincing evidence that they have all evolved from a common ancestor.

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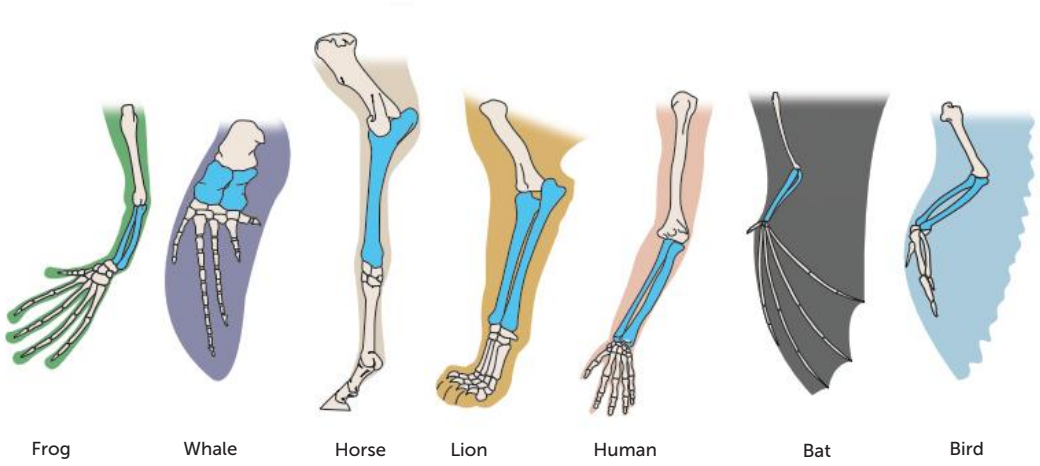

FIGURE 11.14 Left forelimb bones of seven vertebrates; from left to right: frog forelimb, whale flipper, horse forelimb, lion forelimb, human arm, bat wing, bird wing

Anthropoids, the human-like primates, show a great many anatomical resemblances. The number of traits shared and the degree of similarity between the shared traits is remarkable, especially considering the range of habitats occupied. Figure 11.15 shows the skeletons of a human and a gorilla; Figure 11.16 shows the arrangement of muscles in their legs. Note the high degree of similarity between them. These two species share a common ancestor and are therefore closely related. This will be discussed further in later chapters.

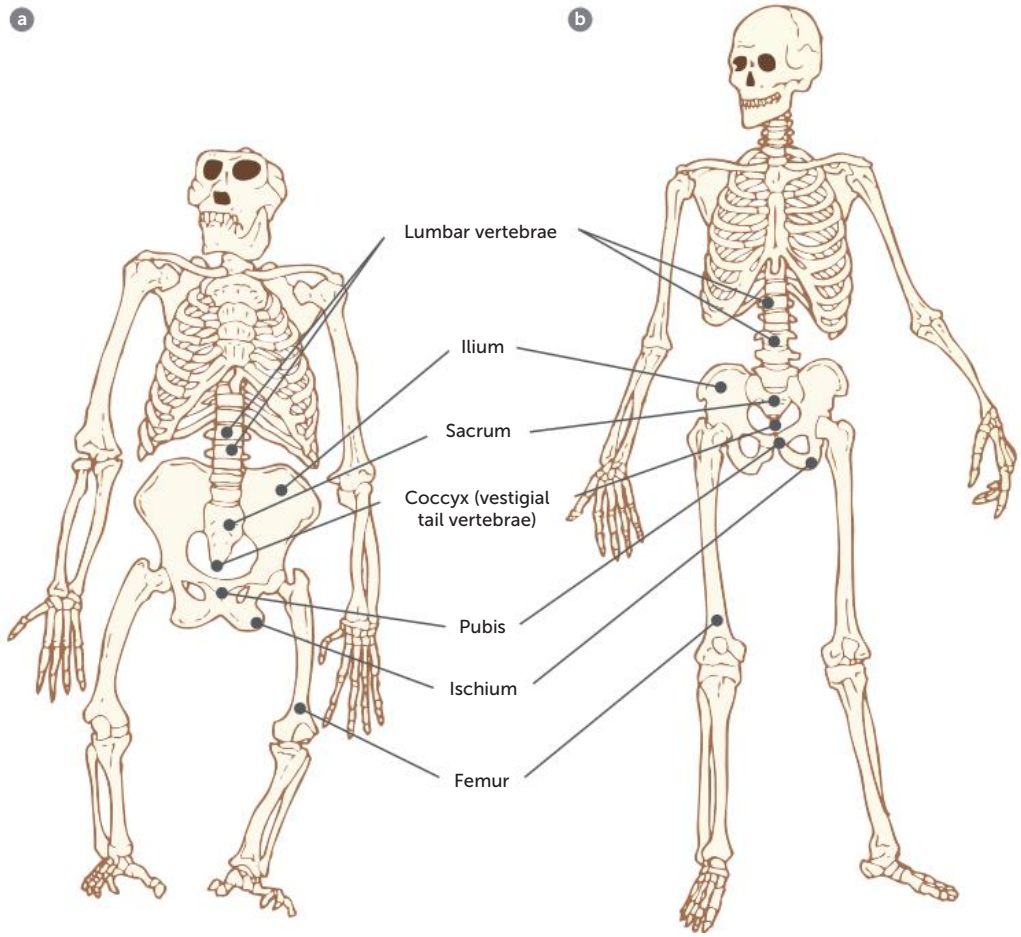

FIGURE 11.15 Skeleton of **a** a gorilla and **b** a human

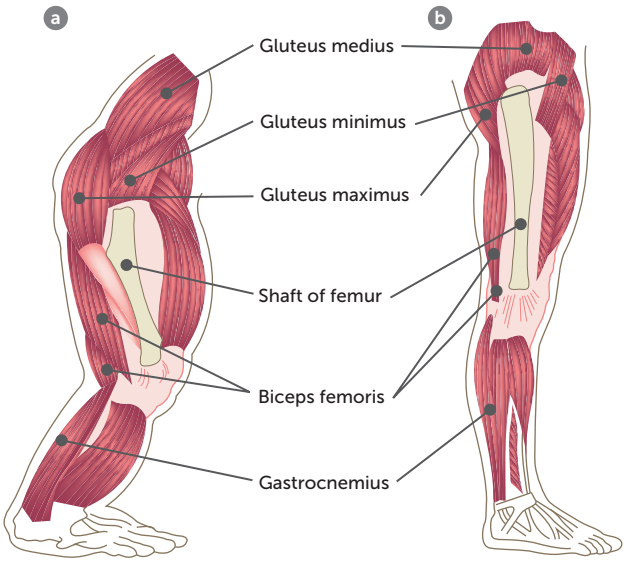

FIGURE 11.16
Musculature of the right leg of **a** a gorilla and **b** a human

Vestigial structures

Vestigial structures are structures that have changed during evolution to the point that they no longer fulfil their origin function. They are common in vertebrate species and form an intriguing aspect of comparative anatomy. Humans may have as many as 90 of these structures. Keep in mind that vestigial structures are largely or entirely functionless when their original role is being considered. However, some may retain lesser functions or develop new ones.

- The **nictitating membrane**, or transparent third eyelid, found in cats, birds, frogs and other vertebrates is able to cover the eye for protection. In humans, it is only represented by a pinkish membrane located at the inner corner of each eye that is unable to cover the eye. Therefore, it cannot fulfil its original function. However, it does have some use in maintaining tear drainage.

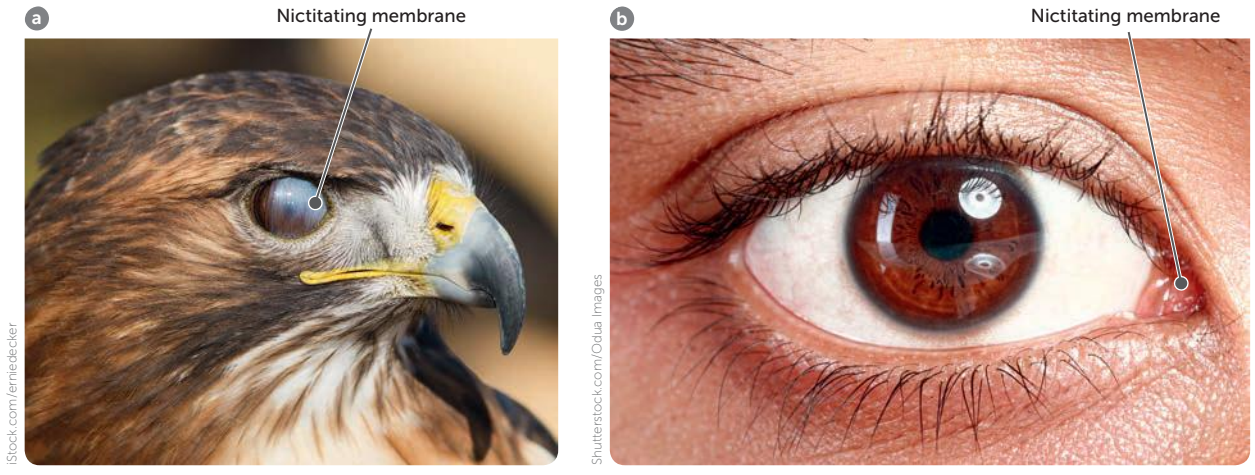

FIGURE 11.17 a The nictitating membrane of falcons protects the eye while diving at high speeds; **b** The nictitating membrane of humans is considered a vestigial structure as it does not fulfil its original function

- The muscles that move the external ears of many mammals are reduced to such an extent in humans that, in most individuals, they will not move the ears at all.

- In most humans the third molars, or wisdom teeth, erupt abnormally and cannot be used in mastication. Frequently, they are removed before eruption so that they do not become painful. About one-fifth of the population are spared any discomfort because the third molars do not develop at all.
- About one-fifth of the population do not develop the muscles that lie above the pubic bone, the pyramidalis muscles. These muscles, if present, do not make any difference to muscular performance.
- Humans still have the vertebrae for a tail, fused to form the coccyx.
- Males have nipples on their chests, although some would argue that these should not be termed vestigial, as they had no function in the first place. They appear to be retained in males because all human foetuses develop from the same basic genetic form, and nipples do have an important function in females.
- The muscles at the base of hairs are considered to be vestigial in humans. In mammals with fur or spines, and in birds with feathers, these tiny muscles pull the hair or feather upright, creating a layer of insulating air to protect against the cold. However, human hair is so fine that it is not capable of such a function and the contraction of the muscles is seen as goose bumps.
- In humans the appendix has commonly been considered a vestigial structure. In herbivorous vertebrates the appendix plays a role in the digestion of tough plant matter. It is thought that it played a similar role in ancient humans, whose diet was more herbivorous. With the change in diets of humans to more easily digested food, the appendix is no longer needed for its original function. Many humans have their appendixes removed following inflammation known as appendicitis. As there does not appear to be any negative effects of the removal, scientists used to think that the appendix does not have any function in present-day humans. However, research is now indicating that the appendix actually has a role in the immune system, and that it produces and stores good gut bacteria. Based on this information, there is now debate about whether the appendix is actually a vestigial structure.

Evolutionary mechanisms can be used to explain the existence of many of these structures that appear to have no function. They are what remain of organs that were functional in ancestral forms. Over time, and with changing environmental conditions, such organs were no longer essential to survival and were gradually reduced to vestigial remnants. As these remnants are not harmful in any way, they have not been completely eliminated. However, natural selection has reduced the organs to non-functional remnants because it would have been a waste of the organism's energy and resources to maintain useless structures. Such organs will probably disappear altogether as there is no selection pressure to retain them.

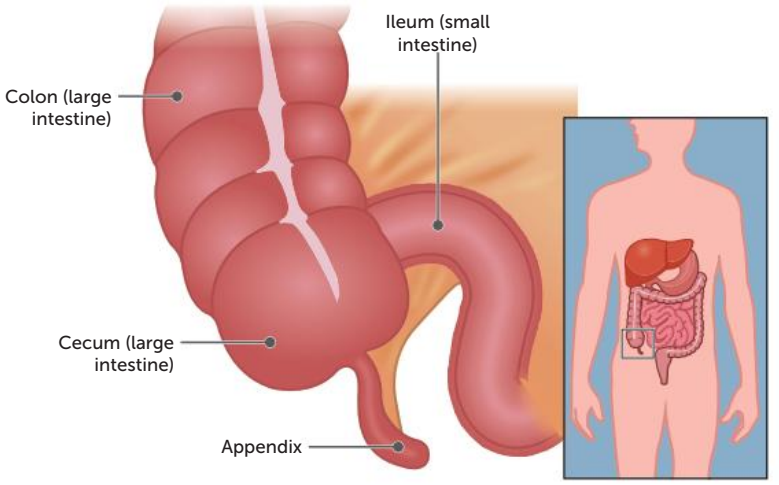

FIGURE 11.18 Location of the appendix

Why do humans have an appendix?
This website has more information about the appendix in humans.

Vestigial organs
This website provides more information about vestigial organs.

7 vestigial features of the human body
This website explains seven vestigial features in humans.

Evidence for evolution
This website provides a good overview of the evidence for evolution.

Key concept

Comparison of the anatomy of embryos, homologous structures and vestigial structures provides evidence for evolution.

WS

11.2 Evidence for evolution

Questions 11.3

RECALL KNOWLEDGE

1. Define 'homologous structures' and 'vestigial structures'.
2. Describe how comparing the structure of embryos at different stages of development can provide evidence for evolution.
3. Describe how a comparison of homologous structures provides evidence for evolution. Include an example in your explanation.
4. List five vestigial structures of humans.

APPLY KNOWLEDGE

5. Explain the importance of fossils in providing evidence for evolution.
6. Explain how our developing understanding of the functions of the appendix has led some scientists to believe that it is not a vestigial structure.

11.4 PHYLOGENETIC TREES

The techniques described in this and the previous chapter enable scientists to work out the likely evolutionary relationships between groups of organisms. The probable relationships can then be represented as a diagram, called a phylogenetic tree. A **phylogenetic tree**, also called a **dendrogram**, represents the evolutionary relationships between a number of organisms derived from a common ancestor. The ancestral organism forms the base of the tree, and those organisms that have arisen from it are placed on the ends of 'branches'. Relationships between the various organisms are shown by the distance between them on the tree, with closely related groups positioned on 'branches' close to one another. However, keep in mind that these are only inferred relationships; different researchers may come up with different 'trees' to fit their interpretations of the data.

Phylogenetic trees are often used to simplify more complex relationships, in order to enable them to be more easily understood. For example, in Figure 11.19 the tree on the left, labelled 'a', shows a hypothetical ancestral population that has divided to form two separate populations; each of these has divided further to produce four descendant species, A, B, C and D. Notice that the branches are thick and curving to represent the variation that may have existed in the past, and that there are side branches that have died out over the period represented. The tree on the right, labelled 'b', is a simplified representation using thin, straight lines to show the lineages of just the four descendant species A, B, C and D.

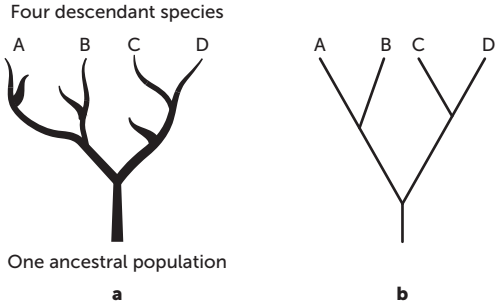

FIGURE 11.19 A simple phylogenetic tree representing one ancestral population and four descendant species

Activity 11.3
Investigating phylogenetic trees

Phylogenetic trees are useful for representing relationships as well as organising knowledge of genetic diversity and structural classifications. They are particularly useful for showing evolutionary pathways and have been used as such since the time of Charles Darwin.

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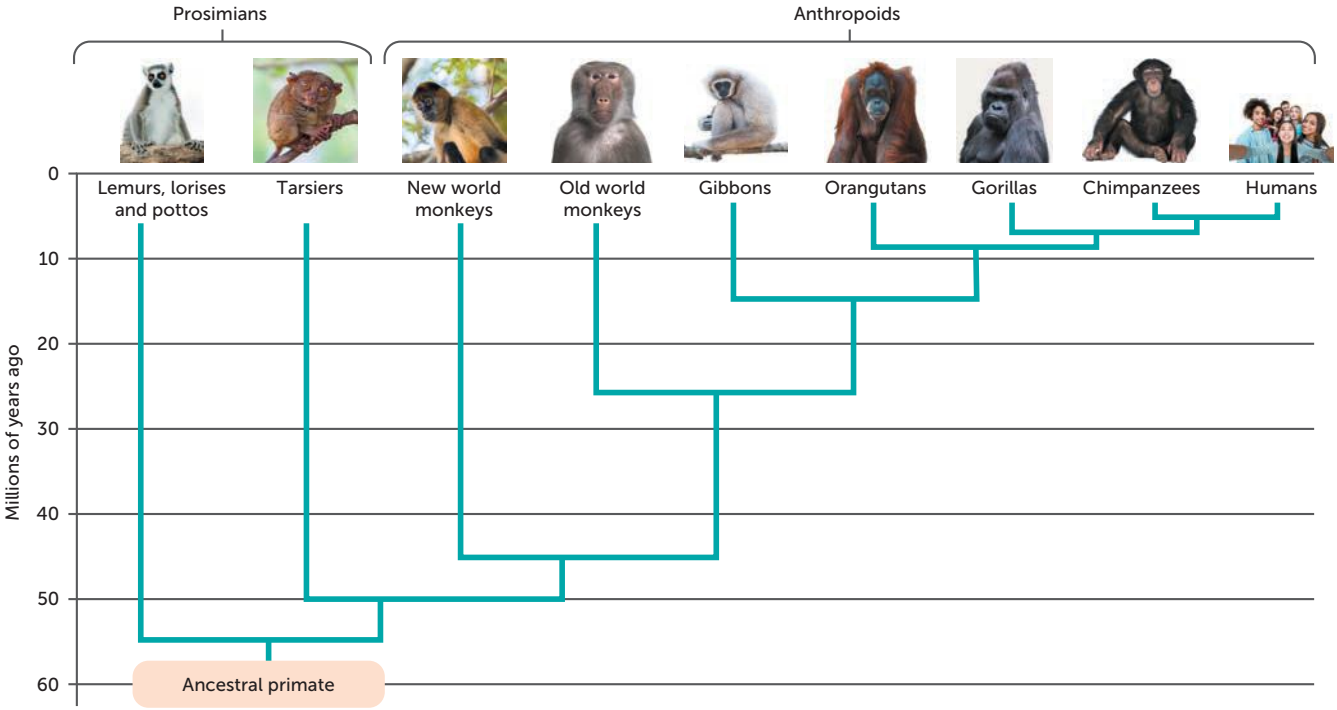


FIGURE 11.20 A phylogenetic tree of primates

Key concept

A phylogenetic tree shows the evolutionary relationship between species, with species with more recent common ancestors diverging further up the tree.

Drawing phylogenetic trees

A variety of information can be used to draw phylogenetic trees, including the presence (or absence) of traits and the number of similarities (or differences) in DNA or amino acid sequences. In all cases, species with the greatest similarities are drawn with branches closer together, while those with the most differences are drawn with branches the furthest apart.

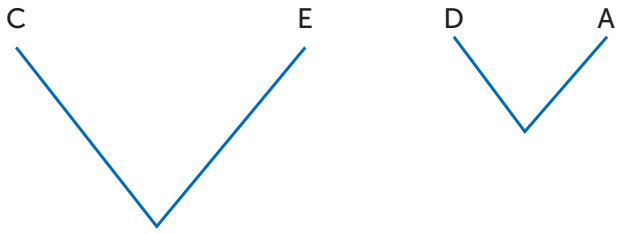
We will use data about the number of differences in amino acids in a protein found in five fictional species, named A, B, C, D and E, to draw a phylogenetic tree. This data is shown in Table 11.2.

TABLE 11.2 The number of differences in amino acids in a protein in fictional species

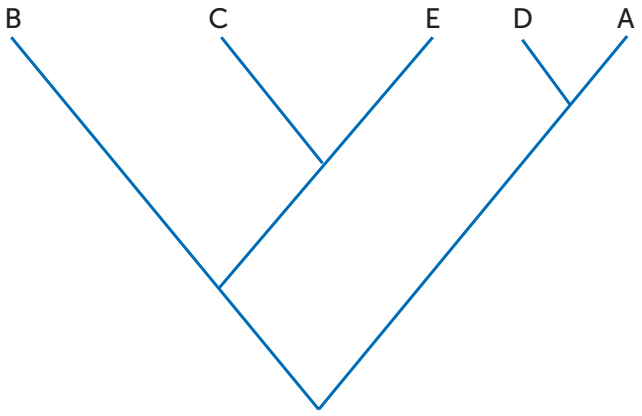
	A	B	C	D	E
A		41	26	1	31
B			15	40	20
C				25	5
D					30
E					

STEP 1: Identify species with the most similarities (A and D). Draw these joined by short branches showing a recent common ancestor.

STEP 2: Identify the species with the next most similarities (C and E). If these are also similar to the species identified in step 1, draw them branching underneath the most recent branch showing the recent common ancestor. If they are not similar to them, join them by branches independent of the others. In this example, C and E are similar to each other, but not A, B or D. Therefore, they are joined independent of the other species. (Note that the branch for C and E is lower than A and D, representing less similarity and, therefore, a more distant common ancestor.)



STEP 3: Identify which of the branches the remaining species is the most similar to. In this example, B has more similarities to C and E than to A and D. Therefore, it has a more recent ancestor with C and E, and so their branches separate higher up than where B branches from A and D.



NOTE: All species that live in the present are drawn level with each other. If the species was extinct the branch would end lower down.

STEP 4: An axis is drawn to show the progress of time, as shown in Figure 11.20.

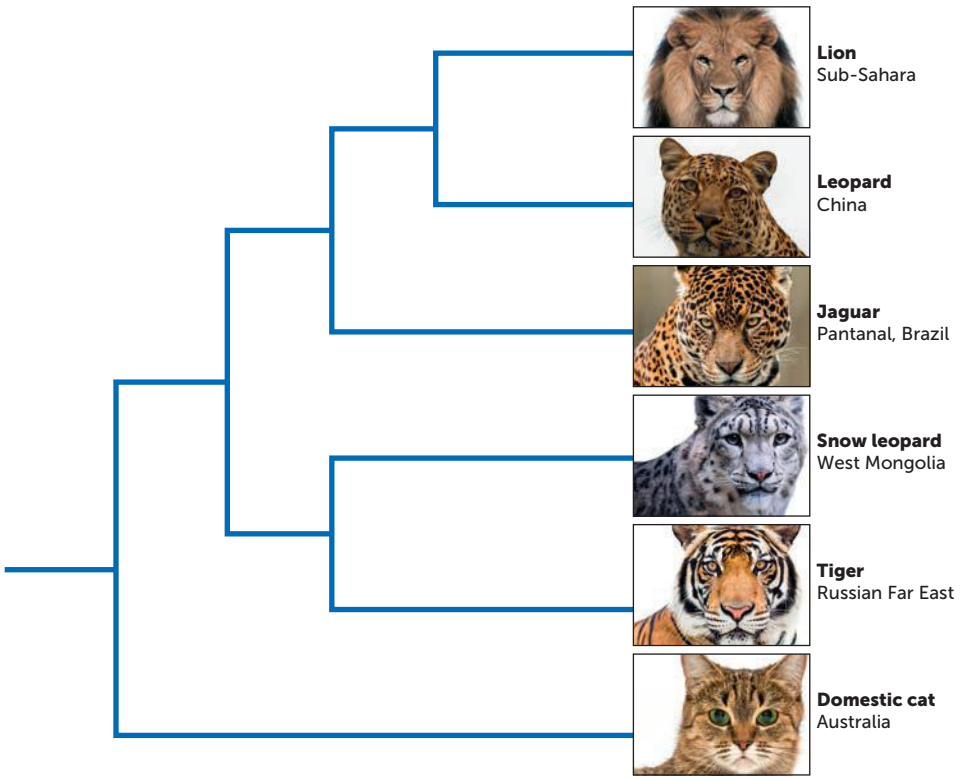
Questions 11.4

RECALL KNOWLEDGE

1 Define 'phylogenetic tree'.
2 Describe what phylogenetic trees are used for.

APPLY KNOWLEDGE

3 Suggest why phylogenetic trees are so named.
4 Consider the phylogenetic tree for cats shown below.
   a Which cats have the most recent common ancestor?
   b Do tigers or jaguars have a more recent common ancestor with lions?
   c Suggest why the domestic cat has more differences than the other cats.



Top to bottom: all Shutterstock.com: Eric Isselee; The Inspired Eye; AP Photographie; andamanec; Suwin; Galyna Andrushko

5 Use the fictional values below comparing four species named A, B, C and D to construct a phylogenetic tree.

SPECIES	PERCENTAGE OF DNA IN COMMON
A and B	80
A and C	65
A and D	62
B and C	75
B and D	72
C and D	98

CHAPTER 11 ACTIVITIES

ACTIVITY 11.1 Investigating radioisotope methods of dating

Radioisotopes are the basis of two of the absolute dating techniques described in this chapter. Isotopes are the different forms of atoms of the same elements. Atoms are composed of particles called electrons, protons and neutrons, and the number of electrons and protons determines the type of atom.

Some isotopes are stable, but other isotopes are unstable, meaning they change into atoms of another element. This is known as decay, and causes radioactivity to be emitted. It is for this reason that they are called radioisotopes.

Radioactive isotopes have a half-life, which is the time it takes for half the atoms to decay, forming atoms of another elements. In the first half-life, half the atoms decay. In the second half-life, half the remaining atoms decay, leaving one quarter of the original material. In the third half-life, half again decay, leaving only one eighth of the original material, and so on.

Radioisotope methods of dating assume that the decay rate of a given isotope is constant and has always been so. Only if nuclear decay rates are constant can the method be used to reliably estimate the age.

You will need

Graph paper; pencils; ruler; eraser

A RADIOCARBON DATING

Radiocarbon dating is based on the decay of the radioactive isotope of carbon, carbon-14 (^{14}C), to nitrogen. When this occurs, radioactivity is released. The decay rate of ^{14}C in material from living organisms or from those that have recently died is 15.6 disintegrations per second per gram of material. After one half-life, 5730 years, the number of disintegrations will be 7.8 disintegrations per second per gram of material (abbreviated to nuclei/s/g).

What to do

1 Draw up a table like the one below and fill in all the gaps.

Decay rate of carbon-14

HALF-LIFE	AGE (YEARS)	RADIOACTIVITY (NUCLEI/S/G)
0	0	15.6
1	5730	7.8
2		
3		
4		
5		
6		
7		
8		
9		
10		

2 On your sheet of graph paper, plot a decay curve for carbon-14 to show the relationship between decay rate and time, up to a maximum of 60 000 years.

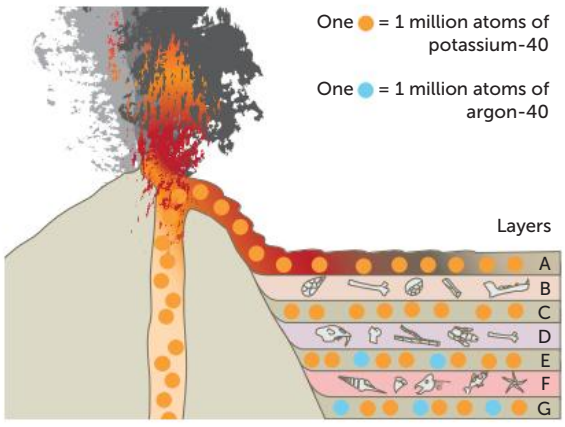
3 Use your graph to answer the following questions.
   - **a** Charcoal remains from a hearth in a cave occupied by Australian Aborigines were found to have a decay rate of 8.9 nuclei/s/g of charcoal. How old was the charcoal?
   - **b** A piece of wood buried in a cave in Europe was found alongside stone tools that were considered to be about 9000 years old. If the wood were the same age as the tools, what decay rate would you expect from the piece of wood?
   - **c** If the piece of wood from question **b** was found to be considerably older than 9000 years, what explanations can you offer for the fact that it was at the same level in the cave deposits as the tools?
   - **d** If the piece of wood was found to be considerably younger than 9000 years, suggest reasons to account for the fact that it was at the same level in the cave deposits as the tools.
   - **e** A fossil bone was discovered and when tested had a decay rate of 1.5 nuclei/s/g. How old was the fossil bone?
   - **f** A piece of fossilised wood was dated using the tree-ring method at 4000 years old. What decay rate would you expect it to display when it was subject to carbon-14 analysis?

B POTASSIUM–ARGON DATING

Potassium–argon dating is based on the decay of radioactive potassium (^{40}K) to form calcium (^{40}Ca) and argon (^{40}Ar). This decay is very slow as potassium-40 has a half-life of around 1300 million years, so the material to be dated must be very old, usually more than 200 000 years. Argon-40 is not normally found in rocks unless it is trapped in solid lava from volcanic eruptions. This occurs when potassium-40 decays. Because the rate at which potassium-40 breaks down into argon-40 is known, it is possible to determine the age of the lava by measuring the ratio of potassium-40 to argon-40.

What to do

Study the following figure carefully, and then answer the questions below. Layers A, C, E and G are lava and contain trapped atoms of potassium-40 and, in most layers, argon-40.



This volcano has erupted periodically for millions of years

1 Explain why there is no argon-40 in layer A.
2 Determine the ratio of potassium-40 to argon-40 in layer E.
3 Rock layers B, D and F are composed of the same material. What type of material do you think this would be? Explain how it has come to be between the alternating layers of lava.
4 Explain why there are no fossils in layers A, C, E and G.

5 Layers B, D and F all contain fossils. For this to have occurred, conditions must have been suitable for fossilisation. Describe the conditions that assist the process of fossilisation.
6 Anthropologists working at this site believe that layer B was formed around 40 000 to 70 000 years ago. This date is too early to use the potassium–argon dating technique. Suggest at least two ways in which they could determine the age of layer B. Explain how each of these methods works.

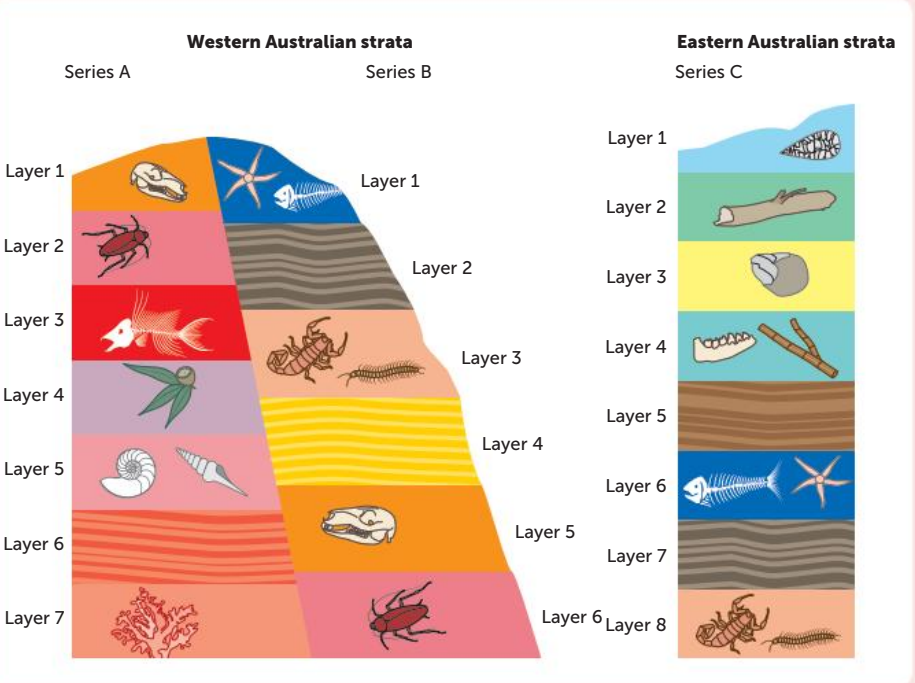
ACTIVITY 11.2 Investigating stratigraphy

Observation of rock strata from various sites around the world has indicated that, in many cases, similar strata contain very similar or identical fossils. In this activity, you will examine three series of hypothetical strata from two locations in Australia. The different strata are shown in three series in the figure below.

What to do

Study the figure carefully and then answer the following questions.

1 How do you think these sediments were formed?
2 The various layers in series A and series B no longer align with each other. Explain how this may have happened.
3 Of all the strata shown in series A, B and C, which is the oldest? Explain how you arrived at your answer.
4 Of all the strata shown, which is the youngest? Explain how you arrived at your answer.
5 Layers A2 and B6, and B1 and C6, contain the same types of fossils. Would these be index fossils? List the criteria that must be met for a fossil to be considered an index fossil.
6 A fossil in layer A4 was dated at 45 000 years using carbon-14 dating. What can you infer about the relative ages of layers B6 and C8?
7 Could layer A6 be dated using the potassium–argon technique? Give reasons for your answer.
8 Do you think dendrochronology could be used to determine an absolute date for layer C2? Give reasons for your answer.



Strata from two locations in Australia

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ACTIVITY 11.3 Investigating phylogenetic trees

The increasing number of techniques in biotechnology is providing scientists with a wealth of data that can be used to examine the evolutionary relationships between species. In this activity, you will use some of this data to construct phylogenetic trees.

What to do

1. Refer to Table 10.2 in Chapter 10 (page 280), which shows the relationship between humans and great apes using DNA differences. Using this information, construct a phylogenetic tree to show diagrammatically the evolutionary relationships.
2. Refer to Table 10.4 in Chapter 10 (page 283). This table shows the differences in amino acids in cytochrome C between humans and a number of other species. The more similarity there is between two molecules, the more recently they have evolved from a common ancestor. Using this information, construct a phylogenetic tree to show the evolutionary relationships between the species listed.
3. Refer to Table 10.5 in Chapter 10 (page 285), which shows the amino acid sequences in the haemoglobin of five mammalian species. Using the data presented in the table, construct a phylogenetic tree to show the evolutionary relationships between the species shown.

Studying your data

Compare the trees you have constructed with those of other members of the class and then answer the following questions.

1. How much variation was there among the phylogenetic trees constructed by different class members? Explain any similarities and differences with the ones you have created.
2. In the three trees you have drawn, does one animal appear to be more closely related to humans?
3. In which of the three trees do you have the most confidence as a good representation of evolutionary relationships? Why did you select this tree?
4. When a phylogenetic tree is constructed, it can be considered a way of presenting a hypothesis. Explain why.

Drawing phylogenetic trees
These websites look at how to draw phylogenetic trees.

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CHAPTER 11 SUMMARY

- Fossils are preserved traces left by organisms that lived a long time ago. They may be bones, shells, teeth, faeces, artefacts or impressions. Fossils give us evidence for evolution.
- Fossils form when parts of organisms become buried quickly in conditions that prevent their decomposition. Therefore, fossils are found in soils near lakes, rivers, caves or volcanoes that are alkaline and low in oxygen.
- Absolute dating gives an actual age of a fossil specimen, while relative dating compares the age of different specimens.
- Potassium–argon dating uses the decay of potassium-40 to form argon-40 and calcium-40. As the decay occurs at a constant rate, the proportion of postassium-40 and argon-40 in a rock allows the age of the specimen to be determined. This method is used for rocks that are older than 200 000 years. Fossils can be dated by the age of nearby rocks.
- Carbon-14 is an absolute dating method that uses the decay of carbon-14 to form nitrogen. When organisms die, the carbon-14 that decays is not replaced as it is in living organisms. Therefore, the ratio of carbon-14 to carbon-12 can be used to calculate the age of the sample. This is useful for samples from living things that are less than about 60 000 years old.
- Stratigraphy is used in relative dating. The principle of superposition states that the layers, or strata, of sedimentary rock on top are younger than those beneath.
- Index fossils, such as some pollen grain fossils, are those that were only present on Earth for a limited time. Therefore, their presence in strata from different areas indicates that the rock in both areas is the same age.
- The fossil record is incomplete, as fossilisation does not always occur; therefore, it is a chance occurrence. It is also difficult to find fossils, as they are buried. Those that are found may be hard to age and may be only part of an organism.
- Fossils and observations of living species allow the anatomy to be compared, providing evidence for evolution by identifying the changes that have occurred.
- Embryological comparisons indicate common ancestors of vertebrates through the presence of slits and arches, a tail, a two-chambered heart and similar brain development.
- Homologous structures – for example, the forelimb bones of vertebrates – have similar structures despite different functions and indicate a common ancestor.
- Vestigial structures have changed during evolution and no longer fulfil the same function. The changes can be used to indicate the degree of separation from common ancestors. Examples in humans are the nictitating membrane, muscles of the ears, wisdom teeth, tails, nipples on males, and muscles at the base of hairs.
- The appendix in humans is often considered a vestigial organ. However, it is now thought that the appendix plays an important role by storing beneficial bacteria and contributing to the immune system.
- Phylogenetic trees, or dendrograms, are used to represent the evolutionary relationships between species. Branches are used to indicate species that have a common ancestor. The closer the branching, the more recent the common ancestor.

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CHAPTER 11 GLOSSARY

Absolute date The actual age (in years) of a fossil or artefact

Accelerator mass spectrometry (AMS) radiocarbon dating A technique used to give radiocarbon dates for very small samples of material

Artefact An object made or modified by humans

Carbon-14 The radioactive isotope of carbon

Correlation of rock strata The process of matching rock strata from different places

Dating Determining the age of excavated artefacts or fossils

Dendrogram *see* phylogenetic tree

Embryology The study of the early development of an organism; in humans, from fertilisation to the end of the eighth week of pregnancy

Fossil Evidence of, or remains of, an organism that lived long ago

Half-life The time required for half of any quantity of radioactive material to decay into stable non-radioactive material

Homologous structure Structures with a similar structure but not necessarily a similar function

Index fossil Fossils or organisms that were on Earth for only a short period of time and are therefore useful in the relative dating of rock strata

Isotope One of two or more atoms of the same element with the same atomic number and number of protons, but different numbers of neutrons

Nictitating membrane A transparent fold of skin (third eyelid) that protects the eyes of birds and reptiles; in humans, it occurs as a vestigial organ in the corner of the eye

Organic compound A compound made up of large molecules that contain carbon

Phylogenetic tree A diagram showing evolutionary relationships between related organisms; also called a dendrogram

Potassium–argon dating A method of calculating the age of a fossil or artefact using the known rate of decay of radioactive potassium

Principle of superposition In a vertical sequence, the principle that the sedimentary rock layers on top will be younger than those lower down

Radiocarbon dating The calculation of the age of a fossil or artefact using the known rate of decay of radioactive carbon

Relative date The age of a fossil or artefact relative to another fossil or artefact (i.e. whether older or younger)

Stratigraphy The study of the sequence of rock layers as a means of relative dating

Vestigial structure A structure of reduced size that appears to have no function; for example, the ear muscles of humans

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CHAPTER 11 REVIEW QUESTIONS

Recall

1 a Define 'fossil'.
b Give examples of five different forms of fossils.

2 a Explain the difference between a fossil and an artefact.
b What is an index fossil? Could there be such things as index artefacts?

3 a What soil types are best for the preservation of fossils?
b Why is it that fossilised soft tissue, such as muscle, is rarely found by those searching for fossils?

4 a What do you understand by the terms 'relative dating' and 'absolute dating'?
b Why is relative dating used when a number of good methods of absolute dating are available?

5 Draw up a table with three columns, listing in the first column the methods of absolute and relative dating described in this chapter. In the second and third columns, list the advantages and limitations of each method.

6 a What is the principle of superposition?
b Does this principle always apply? If not, explain why.

7 What are phylogenetic trees and why are they used?

8 a What are homologous structures?
b Using an example, describe how homologous structures provide evidence for evolution.

9 a What is a vestigial organ? Describe four human vestigial organs.
b Describe the significance of vestigial organs to the theory of evolution.

Explain

10 Explain the principle behind radioisotope methods of dating.

11 Describe why potassium–argon dating cannot be used to date fossil bones.

12 a How is it that the bodies of plants and animals have radioactive carbon-14 in them?
b What does it mean to say that carbon-14 has a half-life of 5730 years?
c Why is it not possible to use radioactive carbon dating on artefacts?
d What is AMS radiocarbon dating?

13 a Explain how index fossils can be used to compare strata from different locations.
b Describe the different ways in which fossil pollen grains can be of use to the anthropologist.

14 How does a study of embryology assist in supporting the theory of evolution? Give examples to illustrate your answer.

Apply

15 Anthropologists excavating the floor of a cave found, at a depth of 50 centimetres, a deposit of charcoal that they concluded was the site of an ancient hearth. Next to the hearth, at the same depth, was a stone tool. Radiocarbon analysis of the charcoal showed that the ratio of carbon-14 to carbon-12 was 0.25 in 10^{12}. Further excavation uncovered, at a depth of 95 centimetres, a fragment of human jaw bone and the thigh bone of another animal.

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a What would be the estimated absolute age of the stone tool?
b What evidence would suggest that the jaw bone and thigh bones were the same age?
c Further testing showed that the thigh bone was younger than the jaw bone. How could this be possible?

16 The sand dunes around the Australian coast consist of alkaline soil. If an animal were buried in the dunes by drifting sand, would its bones become fossilised, provided they were left undisturbed for long enough? Explain the reasons for your answer.

17 In the peat bogs of England, Denmark and other parts of northern Europe, human bodies up to 4000 years old have been found. The hair, skin and other soft tissues have been so well preserved that the fingerprints can still be seen on the skin of the hand, and food in the alimentary canal is complete enough to indicate the nature of the last meal eaten.
a Describe the types of conditions that must be present in peat bogs to allow preservation of these tissues for such a long period of time.
b Would you expect the skeletons of these 'bog people' to be preserved? Why, or why not?

18 Riversleigh, in north-west Queensland, is one of the world's most important and abundant fossil sites. Fossils found at Riversleigh include kangaroos, wombats, bandicoots, possums, koalas, platypuses, crocodiles, snakes, turtles, lungfish, birds, frogs, snails and insects.
a From this list of some of the fossils found at Riversleigh, write a description of what the area must have been like when the fossil animals were alive.
b What conditions must have occurred at Riversleigh for so many organisms to have been fossilised?

19 Homologous organs are so called because they have a similar structure. However, the basic structure may be modified substantially to carry out a different function. Describe the changes that have taken place to the vertebrate forelimb for it to become:
a a flipper
b a wing
c an arm.

Extend

20 In this chapter, the forelimb was used as an example of homologous structures. What other structures found in vertebrates could be used to illustrate homology?

21 In 1893 a German anatomist, Robert Weidersheim, compiled a list of 86 vestigial organs. On his list were the valves in veins, the tonsils, the pituitary gland and the thymus. Why must scientists be very careful about describing an organ as vestigial?

22 More than 135 years ago, Charles Darwin predicted that fossils of the ancestors of modern humans would be found in Africa. Suggest what evidence Darwin would have used as the basis for making that suggestion.

23 New techniques in establishing an absolute age for a fossil or artefact have been developed in recent years. Three of these are uranium–thorium dating, electron spin resonance and thermoluminescence. Find out:
a the principle on which these techniques are based
b the uses to which the techniques have been put
c limitations to the use of these techniques.

12 TRENDS IN HOMINID EVOLUTION

UNIT 4 CONTENT

SCIENCE INQUIRY SKILLS

» conduct investigations, including the use of virtual or real biotechnological techniques of polymerase chain reaction (PCR), gel electrophoresis for deoxyribonucleic acid (DNA) sequencing, and techniques for relative and absolute dating, safely, competently and methodically for valid and reliable collection of data

SCIENCE UNDERSTANDING

Hominid evolutionary trends

» humans as primates are classified in the same taxonomic family as the great apes. The species within the family are differentiated by DNA nucleotide sequences, which brings about differences in:
  – relative size of cerebral cortex
  – mobility of the digits
  – locomotion – adaptations to bipedalism and quadrupedalism
  – prognathism and dentition

Source: School Curriculum and Standards Authority, Government of Western Australia

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12.1 HUMANS AS PRIMATES

Humans, apes, monkeys and some other related animals are called primates because they are all classified together in the taxonomic order Primates. The non-human primates are of special interest to us because they are the closest living relatives of our own species. A comparative study of primates is fundamental to any investigation of the evolution of modern humans. In trying to develop an understanding of how human characteristics evolved, a number of sources of evidence can be used:

- comparative anatomy
- comparative biochemistry, including DNA and proteins
- behaviour of living primates
- fossils of primates.

Humans are classified in the same family as the great apes: chimpanzees, bonobos, gorillas and orangutans. The great apes share the most recent common ancestor with humans and, therefore, share many of our characteristics, including very similar DNA.

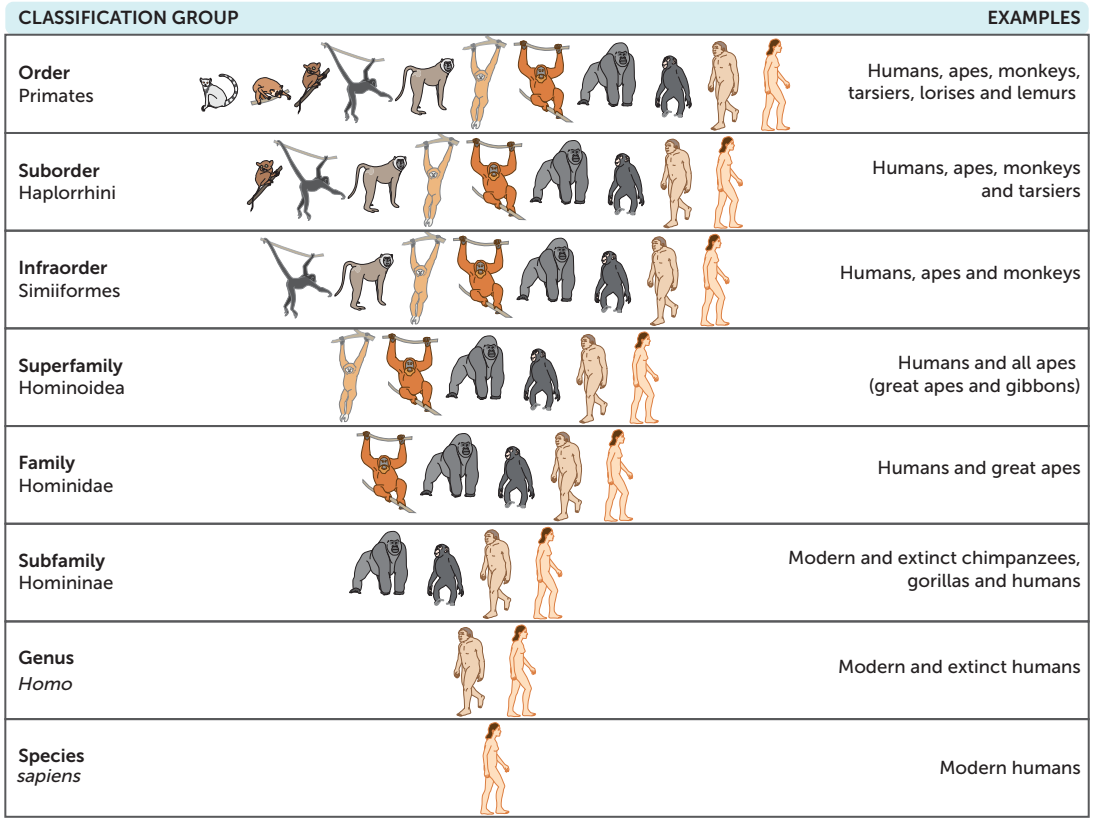

FIGURE 12.1 Diagrammatic representation of the hierarchy within the Primate order

TABLE 12.1 Classification of humans within the Primate order

LEVEL OF CLASSIFICATION	NAME	EXAMPLES
Order	Primates	Primates include tarsiers, lemurs, lorises, monkeys, apes and humans
Suborder	Haplorrhini	Haplorrhini include tarsiers, monkeys, apes and humans
Infraorder	Simiiformes	Simiiformes include monkeys, apes and humans
Parvorder	Catarrhini	Catarrhines include Old World monkeys, apes and humans
Superfamily	Hominoidea	Hominoids include apes and humans
Family	Hominidae	Hominids include all modern and extinct orangutans, gorillas, chimpanzees and humans

LEVEL OF CLASSIFICATION	NAME	EXAMPLES
Subfamily	Homininae	Hominines include all modern and extinct chimpanzees, gorillas and humans
Tribe*	Hominini	Hominins include extinct ancestors of humans and modern humans
Genus	*Homo*	*Homo* includes some extinct ancestors of humans and modern humans
Species	*sapiens*	*Homo sapiens* are modern humans

*Note: *Tribe* is a classification group within a subfamily. The meaning of 'tribe' here is different from its use to describe an ethnic group of people.*

Humans are apes
This website explains why human are apes.

Primate images
This website has some excellent photos of primates.

What is a primate?
This website provides more information about the characteristics of primates.

What makes a primate a primate?
This website describes what makes a primate a primate.

It is common for the term 'ape' to refer to gorillas, orangutans and chimpanzees, and not humans. However, as they are all in the same family, humans can also be considered apes.

Characteristics of primates

There is no one characteristic that can be used to separate the primates from all other mammals. However, some features shared by all primates can be used to identify them as a group. Most of these features are a result of primates having evolved in an arboreal, or tree-like, environment. Two of these, grasping fingers and toes and overlapping vision, when taken together are distinctive to the primates. Table 12.2 summarises the characteristics of primates.

TABLE 12.2 Summary of the characteristics of members of the order Primates

FEATURE	PRIMATE CHARACTERISTICS
Body	Not specialised for a particular environment
Limbs	Generally unspecialised
Hands/feet	Pentadactyl – five fingers or toes Nails instead of claws Grasping fingers and toes with friction ridges for gripping First digit opposable
Eyes	Forward facing for three-dimensional (stereoscopic) vision Most are able to distinguish colour
Sense of smell	Very poor
Teeth	Four incisors in both the upper and lower jaw
Brain	Large and complex Cerebrum size increases as primates become more highly evolved
Reproduction	Not restricted to a breeding season Rhythmical sexual cycle Usually only one offspring at a time Long period of parental care for offspring

Key concept

Humans, chimpanzees, orangutans and gorillas are primates that are classified as great apes.

Questions 12.1

RECALL KNOWLEDGE

1. Monkeys, humans and gorillas are all in which taxonomic order?
2. Define 'arboreal'.
3. List five characteristics of all primates.

APPLY KNOWLEDGE

4. An unknown species is discovered. It has five fingers with claws, an opposable thumb and a small cerebrum that has limited convolutions. Would the species be classified as being a member of the family Hominidae? Explain your answer.

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12.2 VARIATION WITHIN THE FAMILY HOMINIDAE

Humans are in the same family as the great apes (orangutans, gorillas, chimpanzees and bonobos). This family is called Hominidae, and its members are called **hominids**. Previously, only humans were classified in this family. However, advances in molecular techniques have shown that humans share a common ancestor with chimpanzees and gorillas. Orangutans and humans are slightly more distantly related, with bonobos separating at a more distant branch of the phylogenetic tree.

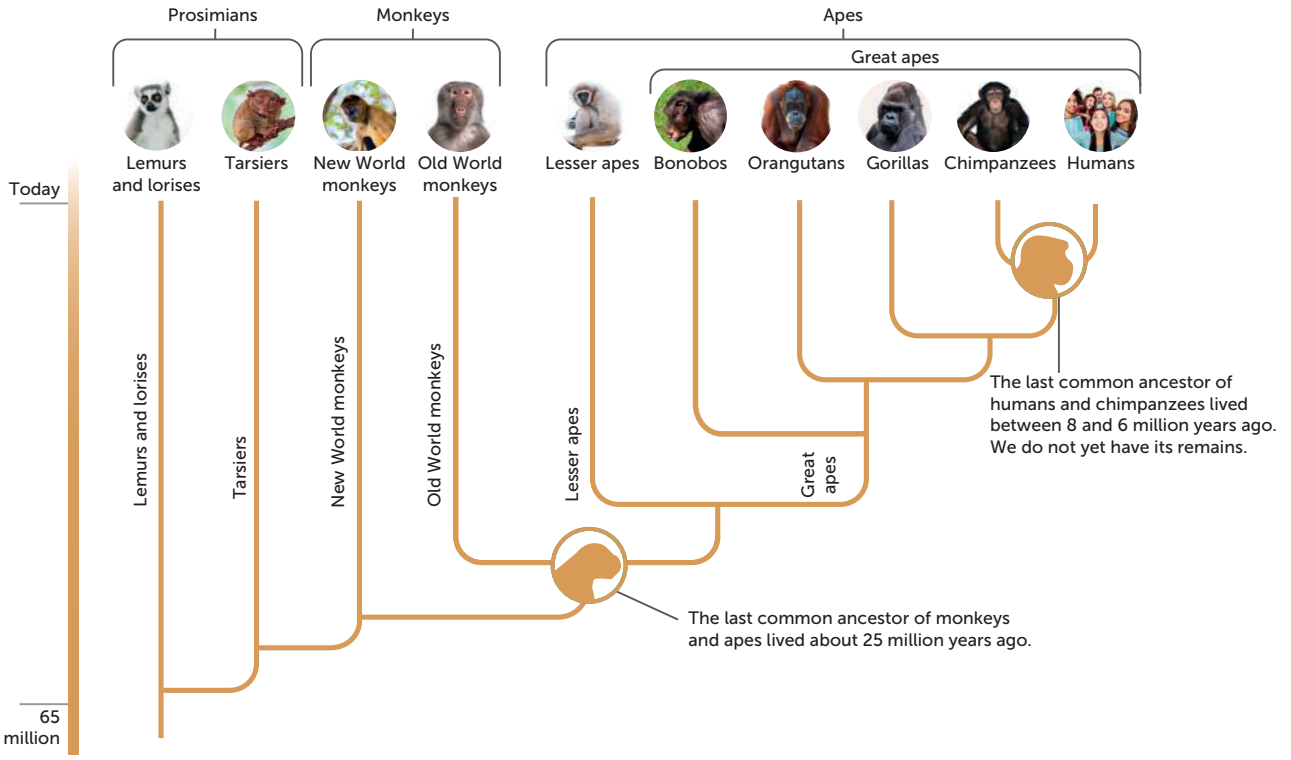

FIGURE 12.2 Phylogenetic tree for primates

All species in the family Hominidae share some characteristics. These include:

- a larger, more complex brain than other primates. This enables an increased cognitive ability that means they can recognise themselves in a mirror
- five cusps in the molar teeth of the lower jaw
- arms that can freely rotate at the shoulder
- a wide, shallow chest cavity
- no external tail
- an appendix
- being active during the day (**diurnal**).

However, there are also some variations that reflect changes in the DNA nucleotide sequences during evolution. Some of these changes relate to:

- relative size of the cerebral cortex
- mobility of the digits
- locomotion – adaptations to bipedalism and quadrupedalism
- prognathism and dentition.

Changes in the relative size of the cerebral cortex

In the primates, the part of the brain responsible for complex functions, the **cerebrum**, has progressively increased in size. This is especially true of the outer region called the **cerebral cortex**. It is this region of the brain that is concerned with so-called higher functions – vision, memory, reasoning and manipulative ability. These functions are necessary to cope successfully with changes in the environment. Figure 12.3 illustrates the increase in size and complexity of the cerebrum from lemurs to humans. This is one of the most significant features of primate evolution.

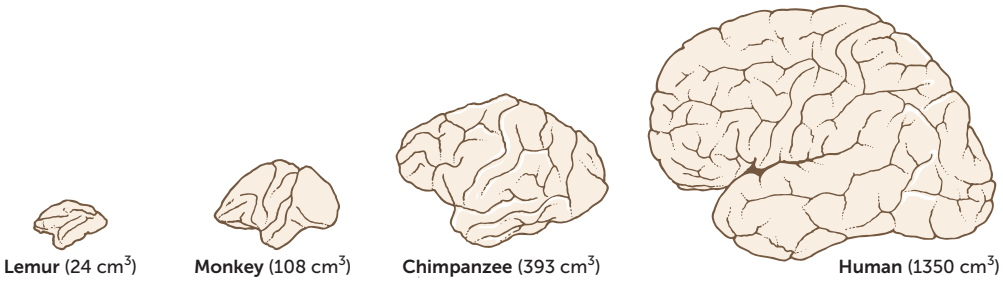

FIGURE 12.3 The increase in size and complexity of the cerebrum in various primates (drawn to scale)

Primates have large brains for their body size. This seems to be a consequence of their tree-dwelling environment. The pressure of natural selection in an arboreal environment would have favoured more accurate visual and tactile perception along with better coordination between such sensory stimuli and any muscular response. Unlike smell or hearing, the reliance on vision to move about, and to locate and manipulate food, generates a large amount of complex sensory information that has to be processed and stored. In primate brains, such operations are carried out by the cerebral cortex. Progressive expansion of the cerebral cortex has resulted in it becoming so large that it covers the rest of the brain. This is most noticeable in humans.

Humans' brains range in size from 900 cm^3 to 2200 cm^3, but average around 1350 cm^3 in adulthood. This contrasts markedly with those of the other apes, which average between 400 cm^3 and 500 cm^3. Most of the difference in brain size is associated with the cerebrum, especially the cerebral cortex.

Compared with other apes, the front part of the cerebrum, known as the **frontal lobe**, has the greatest enlargement in surface area. In humans it makes up 47% of the total cortical surface, whereas in apes it comprises only 33%. It is in the frontal lobe that the higher functions of thinking, reasoning, planning and processing take place.

The brains of hominids have a strong pattern of convolutions. These **convolutions**, or folds, enable the surface area of the brain, and hence the cerebral cortex, to be greatly increased. Notice how, in Figure 12.3, the cerebrum becomes larger and more convoluted as we progress from lemur to monkey, chimpanzee and human. These convolutions have resulted in a 50% increase in the surface area of the human brain compared with what it would be on a brain with no convolutions.

The increase in size of the cerebral cortex has had far-reaching effects on the way primates live. It has enabled them to move about and locate food, and to develop special skills. One of the most significant of these is tool making. This ability is most highly developed in humans, but is also seen in chimpanzees. Tool making, as opposed to tool use, involves a predetermined image of what the completed tool should look like – something only possible with a highly developed brain.

Primate brains
This website shows views of the brains of various primates and other mammals. Click on the particular species and then on the brain photographs to see an enlargement.

In addition, an increase in the size of the cerebral cortex would have allowed a greater variety of behavioural responses to meet a wide array of environmental problems. For most primate species, daily life involves numerous interactions with relatives, allies and adversaries. Mutual cleaning and grooming help to reinforce relationships, while threats, sometimes followed by fighting, maintain the hierarchy of dominance that pervades many primate troops. Such behavioural flexibility has taken the place of further physical specialisation.

A large brain requires a large brain case, or **cranium**, and in humans more of the skull is used in housing the brain than in the other apes. As a consequence, the brow tends to be vertical and lacks the prominent brow ridges possessed by the apes. These features, together with a shortening of the snout, have given humans a flat face, although the bones of the nose still protrude. For this reason, humans have a far more prominent nose than any other primate.

Activity 12.1
Comparing primate skulls

The brains of early **hominins** have not been fossilised, but because the brain fills the whole of the cranium, brain size can be determined by measuring the volume inside the cranium using an **endocast**. This capacity is known as **cranial capacity**.

Mobility of the digits

The limbs of primates tend to be unspecialised in structure, which allows for great diversity in their use. They are **pentadactyl**, which means they have five digits on each limb. The digits are highly mobile, a feature that can be related to the arboreal way of life of primate ancestors. Grasping, or **prehensile**, digits were essential for climbing by wrapping the long, curved digits around the branches of trees.

The evolutionary trend is towards increasing ability to move the digits independently of one another. The most highly developed digits in this respect are the thumb and the big toe. Not only are they independent, but they are also opposable in most primates. **Opposability** means that the first digit can be moved in such a way that it can touch each of the other digits.

The degree of opposability varies from species to species and depends on the relative length of the first digit compared with the other four. Almost all species of primate show some opposability of the big toe, with humans being the one notable exception. Our big toe is not opposable at all. Opposability was lost when the human foot became a weight-bearing, rather than a grasping, appendage. However, humans do possess the longest thumb of all primates and this has contributed considerably to our ability to manipulate objects with our hands.

Having highly mobile digits has enabled humans, more than any of the other primates, to manipulate objects with great skill. The human hand is short and broad, with short, straight fingers and a long, strong thumb, compared

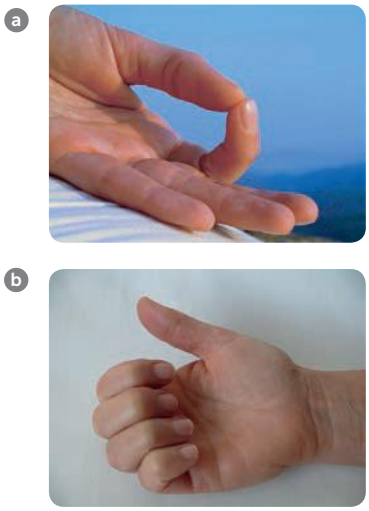

FIGURE 12.4 Manual dexterity of the human hand: **a** Opposability of the thumb as it moves across the palm to touch the other digits; **b** Prehensile digits are capable of being wrapped around an object – here the fingers are being curled towards the palm of the hand

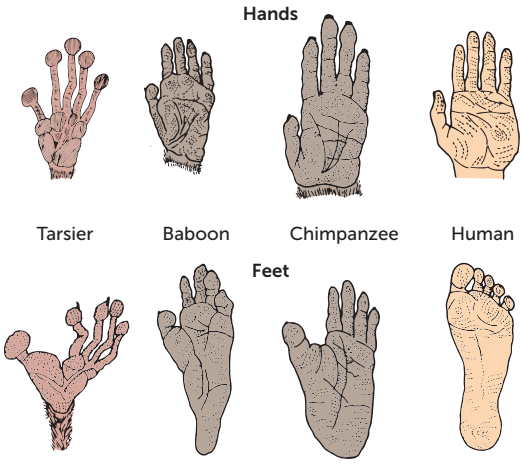

FIGURE 12.5 Hands and feet of four primates. Unlike other primates, humans do not have an opposable big toe

Activity 12.2
Observing the mobility of the human thumb

with that of the other primates. This arrangement gives the thumb a great degree of freedom, and it can readily oppose each of the other digits, thumb tip to fingertip, allowing humans to grasp objects with precision. The **precision grip**, such as that used for holding a pencil when writing, or a needle when sewing, is one of the hallmarks of being human, although it is not unique to humans. What is unique, however, is the amount of contact between the index finger and thumb. This enables humans to handle small or delicate objects effectively. It is different from the **power grip**, which happens when the thumb and fingers apply force against the palm.

Left: Shutterstock.com/Ruslan Kudrin; right: Shutterstock.com/cunaplus

FIGURE 12.6 The **a** power and **b** precision grips

The precision grip requires the presence of a truly opposable thumb and is also seen in Old World monkeys, particularly the ground-living baboons, mandrills and macaques. These monkeys are second only to humans in their manipulative abilities.

Locomotion – adaptations to bipedalism and quadrupedalism

Species in the family Hominidae include humans, chimpanzees, gorillas, orangutans and bonobos. During evolution, there has been a change from **quadrupedalism** (walking on four limbs) to **bipedalism** (walking on two legs). This is a major distinguishing feature of hominins and is used by scientists when classifying fossils.

For humans to be able to stand on two legs and walk bipedally with a striding gait, the skeleton and muscles had to evolve. Compare the skeletons of the gorilla and human in Figure 12.8. The differences seen have evolved over millions of years, so that present-day humans can stand and walk erect on two legs. This **adaptation** helped our human ancestors to survive, and hence has acted as a selective pressure during natural selection.

Clockwise from top left: Alamy Stock Photo/Ger Bosma; Shutterstock.com/EvMedvedeva; Shutterstock.com/wavebreakmedia; iStock.com/SoopySue; iStock.com/007_Bond

FIGURE 12.7 The evolutionary progression from quadrupedalism to bipedalism in **a** bonobo, **b** orangutan, **c** gorilla, **d** chimpanzee and **e** human

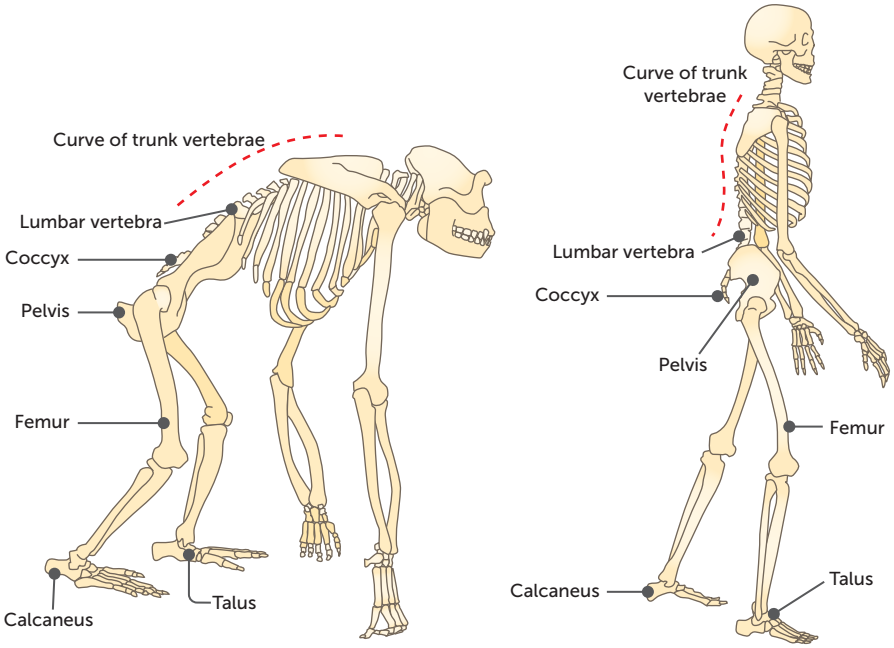

FIGURE 12.8 Posture of the skeletons of a gorilla and a human

Position of the foramen magnum

Where the brain joins the spinal cord there is a hole in the skull called the **foramen magnum**. During the evolution of modern humans from an ape-like ancestor, the foramen magnum has gradually moved forward to become more central. This allows the skull to balance on top of the vertebral column. An ape like a gorilla needs large neck muscles to hold the head in position. In humans, the weight of the skull is borne by the vertebral column and so large neck muscles are not required.

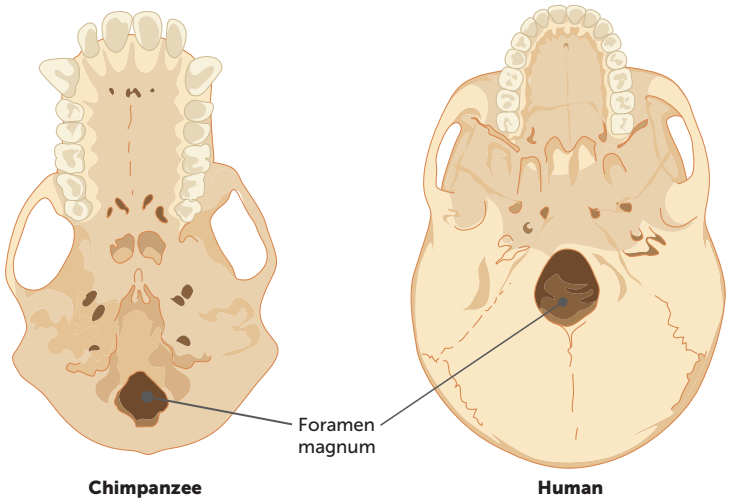

FIGURE 12.9 Base of a chimpanzee skull and a human skull, showing the position of the foramen magnum

Curvature of the spinal column

During evolution, the curvature of the spine has changed to allow an upright posture. The smooth 'C'-shaped curve seen in the spines of apes such as gorillas has evolved to an 'S'-shaped curve in humans. This improves the body balance in the upright position and enables the head to balance on top of the neck.

In humans, the double curvature is achieved by the vertebrae in the lower, or **lumbar**, region being wedge-shaped from front to back, thus forming a forward-jutting curve. In addition, the cervical curve in the neck brings the vertebral column directly under the centre of gravity of the skull.

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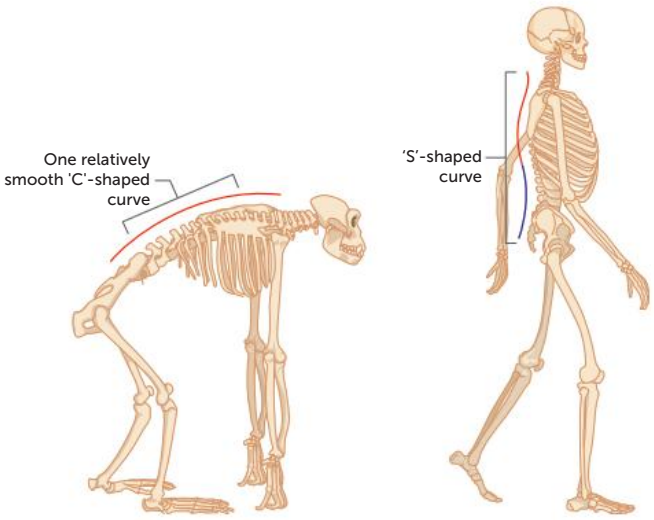

FIGURE 12.10 The progression of a smooth C-shaped curve in a chimpanzee to an S-shaped curve in humans

The jaw

Apes have a protruding jaw, known as **prognathism**, whereas in humans the facial profile is much flatter. During evolution from an ape-like ancestor, the size and protrusion of the human jaw has gradually been reduced. This change has been important in allowing the skull to balance on the top of the spine, because the weight in front of the foramen magnum is approximately equal to the weight behind it. Balance is thus achieved with a minimum of muscular effort.

The pelvis

At its lower end, the vertebral column articulates with the pelvis. The pelvis in humans is broader, and shorter from top to bottom, than in apes, and bowl-shaped (Figure 12.11). This bowl shape supports the abdominal organs when standing erect, provides greater stability for bipedal locomotion and, in the female, supports the developing foetus during pregnancy. The female pelvis tends to be slightly broader than that of the male to allow for the passage of the infant at birth.

The broad hip bones provide space for attachment of the large buttock muscles, which move the legs and keep the upper body erect.

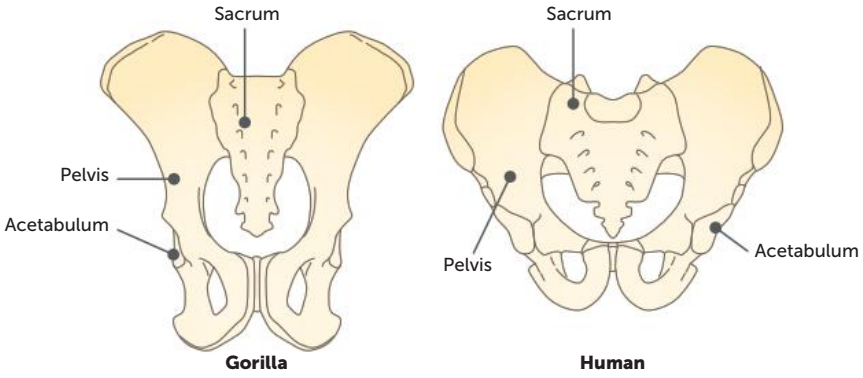

FIGURE 12.11 Pelvises of a gorilla and a human

The carrying angle

In humans, the shape and orientation of the pelvis result in the hip joint being directly under the trunk and head. This allows the weight of the body to be transferred from the pelvis to the legs. The head of the **femur**, or thigh bone, is large and fits into the **acetabulum** (hip socket) of the pelvis. Because the pelvis is broad, the hip sockets are wide apart, but the femurs tend to converge towards the knees. This arrangement of the femurs forms an angle to the vertical, termed the **carrying angle**, which ensures

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that weight distribution remains close to the central axis of the body when walking. As Figure 12.12 indicates, in humans the weight tends to fall through the outside of the femur, whereas in other apes the reverse is true.

The carrying angle allows for greater stability in an upright posture. When walking, it enables the body to be rotated about the lower leg and foot, and each footstep follows a more-or-less straight line. This enables humans to have a striding gait instead of swaying from side to side as do gorillas or chimpanzees when walking on two legs.

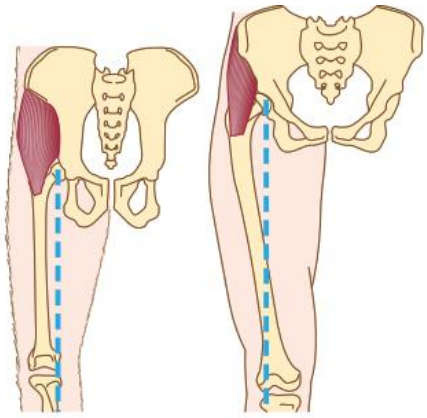

FIGURE 12.12
Pelvises and femurs of chimpanzees and humans, showing how humans have a carrying angle, with the femur angled in towards the knee (the dotted line shows the direction of weight transmission)

The knee

In bipedal species, the weight of the body is transmitted down the outside of the femur to the knee. The knee joint is a two-part hinge joint, with one 'hinge' on either side of the ligaments in the middle of the joint. Because the weight is transmitted to the outer 'hinge', it is larger and stronger than the inner one. Although the weight of the body is transmitted down the outside of each leg, the centre of gravity of the body tends to fall through a line just in front of the knees. This results in a force that tries to bend the knee backward but is resisted by the ligaments making up the knee joint. This natural resistance produces a joint that requires no energy to support the body in a standing position.

The foot

From the knee joint, most of the weight of the body is transmitted through the tibia to the ankle. The tibia is the larger and stronger of the two lower leg bones. At the ankle, body weight is transmitted from the tibia through the talus (ankle bone) to the other tarsal bones, then to the metatarsals and phalanges via the arches of the foot.

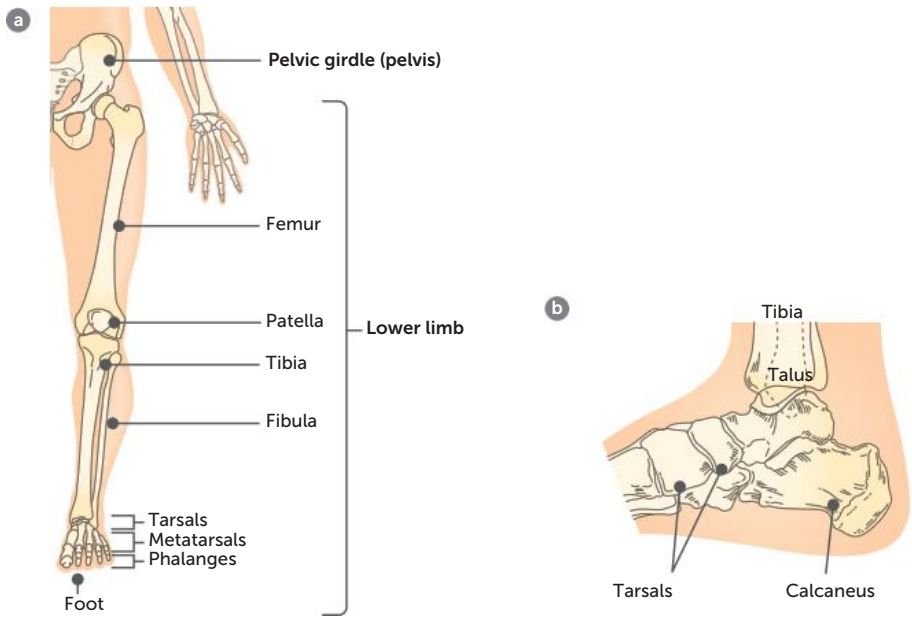

FIGURE 12.13
a Lower limb showing the weight-bearing bones; **b** Tarsal bones of the foot

The human foot is one of the most distinctive adaptations for bipedal locomotion. In becoming a highly specialised locomotory organ it has lost its grasping ability, or prehensility. This is most noticeable with the big toe, which in humans is quite large and aligned alongside the other toes. The bones of the foot between the toes and the ankle, the **metatarsals**, are shaped in such a way that they form two arches: a **longitudinal arch** running from front to back, and a **transverse arch** running from side to side.

The transverse arch is unique to humans. These two arches have enabled humans to perfect bipedal locomotion.

Centre of gravity

Unlike other apes, humans have legs that are longer than the arms. The relatively long legs increase the length of the stride when walking. Surprisingly, they also serve to lower the centre of gravity of the body, the point at which all the weight of the body appears to be concentrated. In contrast to humans, where almost half the total height is in leg length, in apes only about one-third of the total height is taken up in leg length. This results in their centre of gravity being further up the body. Whereas the centre of gravity for the ape is at chest level, for humans it is at the level of the pelvis. The lower centre of gravity in humans contributes to stability when moving bipedally or when standing erect.

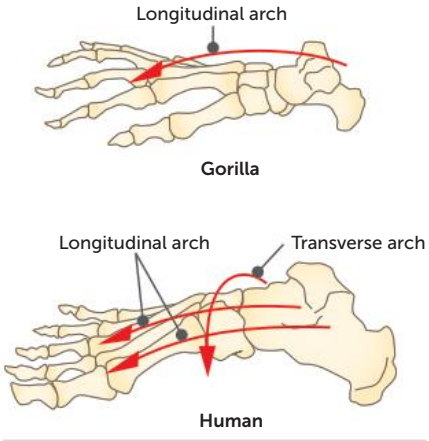

FIGURE 12.14 Arches of the foot of a gorilla and a human: humans have a longitudinal arch and a transverse arch; gorillas have only the longitudinal arch

Muscle tone

One of the essential elements for maintaining an upright stance is muscle tone. **Muscle tone** is the partial contraction of skeletal muscles. For example, to keep the head erect and stop it from slumping forward on to the chest, the muscles in the back of the neck are partially contracted; that is, they have tone. If someone falls asleep while sitting up, the decrease in tone is evident as the head nods until the chin is close to the chest.

Sustained muscle tone is most evident in those muscles that support the body in an upright position. In humans, the muscles that do this are those that bring about movement of the spine, hip, knee and ankle, and also the abdominal muscles. The nervous system and a variety of sense organs work together to maintain the tone in these muscles and the equilibrium of the body.

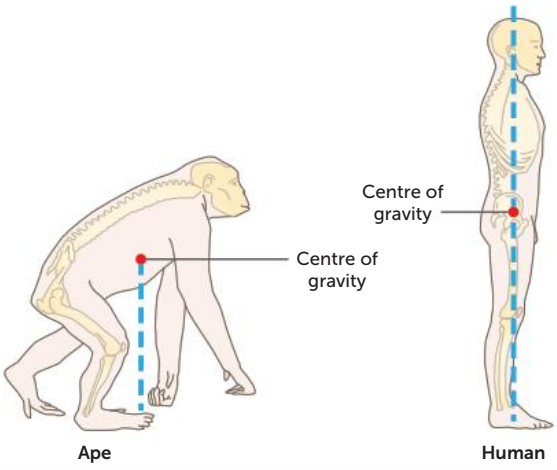

FIGURE 12.15 Centre of gravity of an ape and a human

Walking on two legs – bipedalism
This website has more information about bipedalism.

TABLE 12.3 Summary of the main hominin adaptations for erect posture

STRUCTURE	ADAPTATION
Foramen magnum	Located centrally in the base of the cranium
Jaw bone	Small and non-protruding, enabling the skull to balance on the vertebral column
Vertebral column	Lumbar vertebrae wedge-shaped, producing an 'S'-shaped curve that brings the vertebral column directly under the centre of the skull
Pelvis	Broad; shallow from top to bottom. Provides support for the abdominal organs. Attachment of femurs is wide apart, contributing to the carrying angle
Femurs	Large head of the femur contributes to carrying angle
Knee joint	Outer 'hinge' larger and stronger, to take weight of body. Knee is able to be straightened
Legs	Longer than arms, contributing to a low centre of gravity. Carrying angle allows the weight of the body to be kept close to the central axis
Foot	Large heel bone and aligned big toe form a pedestal on which the body is supported. Foot has both longitudinal and transverse arches
Muscle tone	Partial contraction of muscles to support the spine, hip, knee and ankle

WS
12.1 Adaptations for erect posture

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Key concept

Evolution has led to changes that allow bipedalism in humans. These changes include a central foramen magnum, 'S'-shaped spine, non-protruding jaw, broad pelvis, carrying angle of the femur, knee that is larger on the outside, long legs, longitudinal and transverse arches on the feet, and muscle tone.

Striding gait

Walking upright in such a way that the hip and knee are fully straightened is referred to as the **striding gait**. Hominins are the only animals that have perfected this form of locomotion. Even when walking on their hind legs, the other apes have their knees bent and their bodies bent forward at the hips.

In the striding gait, when the foot hits the ground, weight is transmitted from the heel along the outside of the foot as far as the ball, crosses the ball of the foot (via the transverse arch) and is finally borne by the big toe. At the final moment of striding, the whole weight of the body is propelled by the big toe. This is why the hominins lost the opposability of the big toe; the human foot has evolved into a weight-bearing appendage, rather than a grasping one.

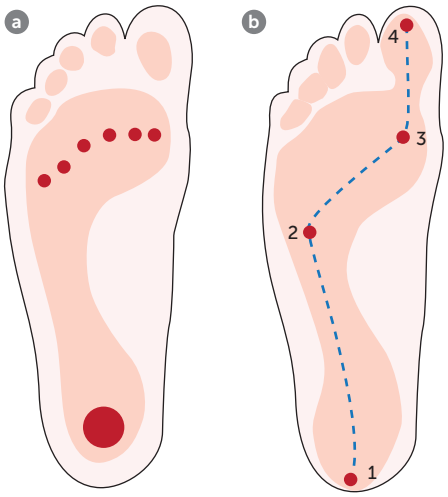

FIGURE 12.16 a How body weight is borne by the foot when standing still; **b** The distribution of body weight as a step is taken: the weight of the body is progressively borne on points 1 to 4 as the heel of the foot hits the ground and the big toe thrusts off

When walking, the trunk rotates about the pelvis. The forward swinging of the arms compensates for this natural rotation of the body: the right arm naturally swings forward as the left leg is extended, and vice versa. Swinging of the arms tends to keep the shoulders at right angles to the direction of travel, and reduces the amount of energy expended. If the arms did not move as they do, energy would be wasted in reversing the rotation of the body after each stride.

In the discussion of the carrying angle earlier in this chapter it was shown that, although the human pelvis is broad and the hip sockets are wide apart, the femurs converge towards the knees. This arrangement of the femurs ensures that weight distribution remains close to the central axis of the body during walking. The arrangement also allows for stability during walking, as the body can be rotated about the lower leg and foot, thus allowing each footstep to follow a more-or-less straight line. Apes such as chimpanzees lack a wide pelvis and carrying angle. When walking on two legs they must sway from side to side so that the body weight is over each leg in turn.

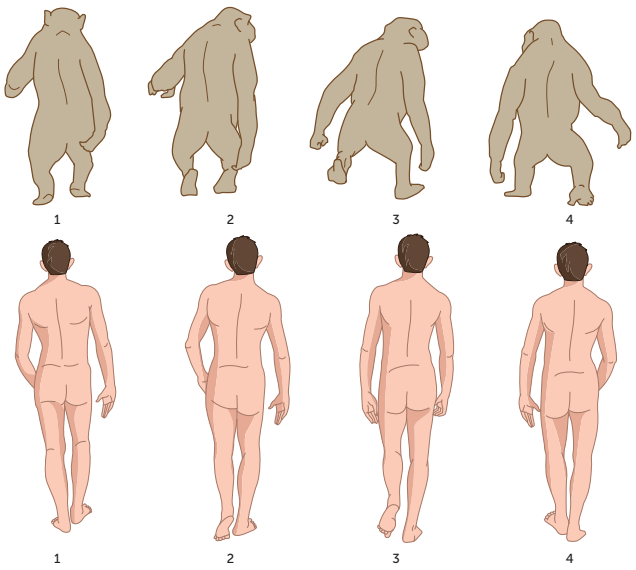

FIGURE 12.17 Comparison of the bipedal walking of a chimpanzee and a human

Advantages of bipedalism

Evolutionary trends arise due to a selective advantage leading to natural selection. In the case of bipedalism, there are a number of possible advantages, including:

- It is a more energy-efficient means of moving.
- It leaves the hands free to use tools.
- It leaves the hands free to carry items.
- The upright stance achieves greater height and thus the ability to see further.
- The upright stance means that less of the body is exposed to sunlight.
- The upright stance increases exposure to breezes, increasing cooling mechanisms.

Activity 12.3
Investigating upright stance and the striding gait

Key concept

Humans have a striding gait due to the carrying angle of the femur and the weight-bearing ability of the big toe. This allows humans to have their weight on one foot without twisting or swaying.

Prognathism and dentition

Evolutionary changes have taken place in the dentition of the primates, in both the number of teeth and in their structure. As with most mammals, primates have two sets of teeth, deciduous (also known as baby teeth) and permanent, and teeth of different shapes that perform different functions.

Number and shape of teeth

The number of each type of tooth that a species has can be expressed as a **dental formula**. The formula gives the number of each type of tooth in one quarter of the jaw. Primitive mammals had a dental formula of 3:1:4:3. This means that there are 44 teeth: three incisors, one canine, four premolars and three molars on each side of each jaw.

Natural selection has resulted in a decrease in the number of teeth in primates compared with early mammals. This is probably related to the gradual reduction in the size of the face and jaw that has occurred in primates that allows the skull to balance during bipedalism. Old World monkeys, apes and humans all have 32 teeth and a dental formula of 2:1:2:3; however, there is considerable difference between them in the structure and arrangement of the teeth.

In the Old World monkeys and apes, the canines are usually large and sharply pointed, projecting beyond the level of the other teeth. Such large canines have required modifications to adjacent teeth

FIGURE 12.18 a The diastema is a gap between teeth that allows the teeth on the opposite jaw to fit in; **b** A jaw without a diastema has the teeth close together

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so that the mouth can be closed. Most primates with large canines have a gap, or **diastema**, between the upper second incisor and the upper canine to accommodate the large lower canine. To allow for the large upper canine, the crown of the first lower premolar is slanted back and has a sharp edge. The upper canine fits tightly against this premolar and is sharpened by the grinding that occurs.

The surface of the molars of apes and humans has evolved from the three cusps of early mammals to four cusps on the upper molars and five cusps on the lower ones. This pattern has been useful in identifying the teeth of fossil apes and humans, and is presumed to have evolved due to the predominantly fruit diet of the apes.

Compared with other primates, human dentition is very distinctive. In humans the canine teeth do not project beyond the level of the other teeth and interlock, as they do in the Old World monkeys and apes. They are more even in size, looking more like incisors. These small canine teeth and relatively small incisors take up less room in the jaw. As a consequence, the shape of the tooth row, or **dental arcade**, has evolved into a different shape. Instead of the 'U' pattern of the apes, it has become parabolic in shape, as shown in Figure 12.20.

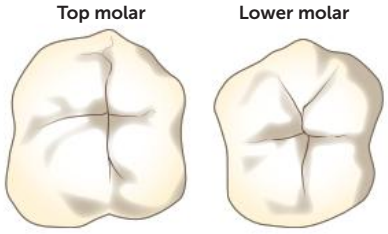


FIGURE 12.19 The difference in structure between a four-cusp and a five-cusp tooth. The 'valleys' between the four cusps of the top tooth form a '+' pattern, while the 'valleys' between the five cusps of the lower tooth form a 'Y'

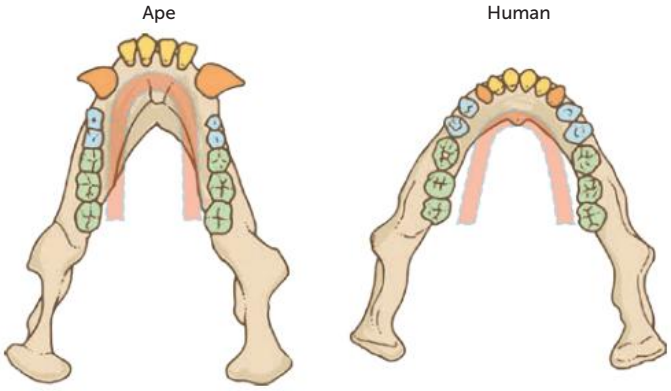


FIGURE 12.20 Dental arcade of a non-human ape and a human

Primate teeth
This website provides more information on primate dentition.

Prognathism and brow ridges

Non-human apes and the early hominins have a forward-jutting jaw, a characteristic known as prognathism, and a distinct **brow ridge**, the bony ridge located above the eye sockets, very evident in adult gorillas. With evolution, the tooth size gradually decreased leading to a flattening of the face, development of a chin and a prominent nose. With the move to bipedalism, a flatter face shifts the weight to a more central position, allowing the skull to balance during an upright stance. And as the size of the frontal lobe has increased, the brain occupies a larger volume, extending the cranium forward and reducing the brow ridges.

Alamy Stock Photo/Sabena Jane Blackbird

FIGURE 12.21 Prognathism is the forward protrusion of the jaw

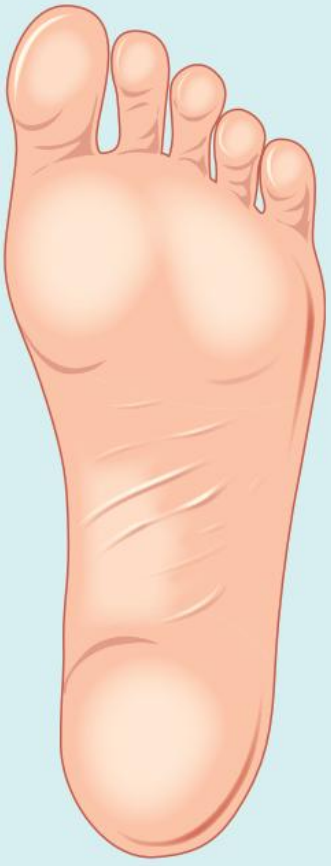
Key concept

During the evolution of primates, the diastema has been lost, the number of teeth and prognathism have decreased, the number of cusps on the molars has increased, and the jaw has become more parabolic in shape.

Questions 12.2

RECALL KNOWLEDGE

1 What family are humans in?
2 List six characteristics of hominids.
3 Describe the trend in the cerebral cortex that has occurred during evolution.
4 Define 'pentadactyl' and 'opposable'.
5 Use an example to describe:
   a precision grip
   b power grip.
6 What is meant by 'bipedal with a striding gait'?
7 List the features that allow humans to have a striding gait.
8 Explain why a bowl-shaped pelvis in humans has an advantage for bipedalism over the longer pelvis of other apes.
9 Define 'carrying angle' and explain why it allows a striding gait.
10 Which side of the knee is larger? Explain why.
11 Describe the distribution of weight from the hip to the foot of a human.
12 Draw the transverse and longitudinal arches on the diagram of a foot below.



13 Describe the trend in prognathism during evolution.
14 State the dental formula of hominids.

APPLY KNOWLEDGE

15 Explain the significance of an increase in the size of the frontal lobe.
16 Which animal would have a more convoluted cerebrum – an orangutan or a chimpanzee?
17 Explain the difference between the size of the brain and the cranial capacity.
18 Explain how the length of the thumb of humans reflects a greater degree of evolution.
19 The spine of humans is described as 'S' shaped. Explain why this is necessary for bipedalism.
20 Explain why doctors are reluctant to amputate the big toe.
21 Gorillas have a diastema, but humans do not. State what a diastema is, and explain why it is present in the jaw of gorillas but not humans.
22 Explain why bipedalism would have allowed 'survival of the fittest' during natural selection.

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CHAPTER 12 ACTIVITIES

ACTIVITY 12.1 Comparing primate skulls

In this activity, you will use a website to compare a number of primate skulls, observing trends in the size and shape of the skull and teeth as one goes from the lemurs and monkeys to apes and humans.

What to do

1. Go to the weblink 'eSkeletons'.
2. Select 'Comparative Anatomy' from the menu at the top. This will enable you to compare two different species of primate.
3. In the comparative anatomy screen, select 'Adult Male Baboon' and 'Adult Male Orangutan' from the Specimen column; then from the Bone column select Cranium, and select 'Lateral' from the View column.
4. You should now have the lateral (side) view of an orangutan and a baboon skull next to each other to compare. Click on the + sign above the images to increase their size.
5. Look carefully at the two skulls, noting the scale listed for each, and take this into consideration when answering the following questions. For some questions it may help to go back and select other views of the skulls for comparison.
   - a Using the scale provided, estimate the length of each skull.
   - b Which skull has a more rounded profile?
   - c Estimate the length of the cranium of each skull. Which species would have the larger and more complex brain? Give reasons for your answer.
   - d Identify and count the teeth that are visible. What is the dental formula for each species?
6. Repeat steps 3 to 5 so that you can compare:
   - a an orangutan with a gorilla
   - b a gorilla with a chimpanzee
   - c a chimpanzee with a human.

eSkeletons

Studying your observations

Review your answers to the questions and use the information collected to describe evolutionary trends in the size and shape of the skull and teeth from baboons to humans.

ACTIVITY 12.2 Observing the mobility of the human thumb

Apes and humans have very mobile digits, but only humans can grip an object with true precision. The human hand differs structurally and functionally from that of the other primates. A longer, stronger thumb that can readily oppose each of the other digits enables humans to manipulate objects using a precision grip. Humans are also able to use a power grip, where an object is grasped between the undersides of the fingers and the palm of the hand, with pressure in the opposite direction being applied by the thumb. We use a power grip when holding a hammer.

In this activity, you will compare the two main ways in which humans use the thumb and fingers to grip objects. Manipulation of objects with both power and precision enabled our ancestors to become efficient tool makers.

You will need

A short length of broom handle or a ruler; a pencil or pen

What to do

1. Hold your hand out in front of you with the back of your hand towards your face. Observe how the position of the thumb is different from the fingers.
2. Move your thumb across the palm of your hand to touch each of your fingers in turn. Note the movement of the thumb.
3. Use your thumb and fingers to pick up a pen or a pencil from your table and hold it as though you are going to write. Observe the way in which the thumb and fingers are employed in the grip you used. This is the precision grip.
4. Using this grip, squeeze the pen tightly and note which muscles are in use.
5. Grasp a length of broom handle or a ruler as you would a hammer. Observe the differences in the position of the thumb and fingers when this method is used to hold an object.
6. Squeeze the broom handle tightly and note which muscles are used. This is the power grip.

Studying your observations

1. In relation to the palm of your hand, how is the position of the thumb different from the fingers? Give two reasons to explain the advantage of the thumb in this position.
2. What term is used to describe the movement of the thumb when it touches each fingertip in turn?
3. a Describe the position of your thumb and fingers when picking up a pen.
   b Which muscles were used to hold the pen in this precision grip?
4. a List the differences in the position of the thumb and fingers when using the precision grip and the power grip.
   b How did your thumb assist in holding an object in the power grip? Describe how it did this.
   c Which muscles were employed in the power grip? Were these different from the ones used in the precision grip?
5. Which of the two grips would be the most efficient at holding an object against force?
6. List the features of the thumb that make both the power and precision grips possible.

ACTIVITY 12.3 Investigating upright stance and the striding gait

A striding gait is a form of locomotion that distinguishes humans from the other living primates. Its evolution depended on changes to the skeleton and associated muscles and joints. In this activity, you will examine some of these features to gain a greater understanding of the way we move.

You will need

A model of a human skeleton; charts or diagrams of the skeleton; the skull of an ape; reference to some of the diagrams in this book. If you wish, you could do the whole activity by comparing the human skeleton with that of the chimpanzee or gorilla at the weblink from Activity 12.1.

What to do

Answer the questions below. As you answer them, refer to the model of the human skeleton, ape skull and figures in the text as directed, or to images on a website.

Studying your observations

1. Compare the skull of an ape with that of a human. List the differences in the size and shape of the crania (brain cases).

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2 Locate the position of the foramen magnum. Look at the base of each skull and compare the position of the foramen magnum in the ape and in the human. Where is the foramen magnum in the human skull? Where is the foramen magnum in the ape skull?
3 Which skull is most easily balanced on the vertebral column: ape or human?
4 Look carefully at the model of the skeleton, and then refer to Figure 12.10. Describe the curves of the vertebral column of the ape and the human. What extra curve exists in the vertebral columns of humans?
5 Look at Figure 12.11 and compare the shape of the human pelvis with that of the gorilla. Which pelvis is wider? Which is longer? Suggest reasons for the relatively wide pelvis in humans.
6 The human pelvis is tilted forward and curves inward, creating a basin shape. List the advantages this arrangement has for upright stance.
7 Look carefully at the model of the skeleton again, and then refer to Figure 12.12. The narrow pelvis of the ape makes the legs hang vertically. This means the ape must keep its feet apart when standing and, when walking, sway from side to side to maintain balance. Describe how the breadth of the pelvis contributes to the carrying angle of the femurs.
8 Explain the effect of the carrying angle on the arrangement of the knees, lower limb bones and the position of the feet in humans. What advantage does this arrangement have for a human walking?
9 The vertebral column of humans acts as a weight-supporting column. How does the shape of the lumbar vertebrae contribute to the lumbar curve? Look closely at the angle between the lumbar curve and the pelvis. What effect does the lumbar curve have on the position of the trunk and legs in humans?
10 Refer to Figure 12.15 and compare the position of the centre of gravity in humans and apes. Which animal has the lower centre of gravity relative to body size? What features of the skeleton contribute to this difference?
11 Describe the pathway the body weight in humans follows from the pelvis down to the feet.
12 Remove your shoe and run your fingers over the top of your foot from little toe side to big toe side. Can you feel the transverse arch? How is this arch different from the longitudinal arch? What is the main function of the two arches?
13 Look at the model of the skeleton again, and then refer to Figure 12.14. Compare the toes of a gorilla and a human. What differences can you see?
14 When humans stride, the big toe provides the thrust. What features of the big toe assist this? Would an ape be able to use the big toe in a similar way? Explain your answer.
15 Describe how the arches of the foot enable weight to be distributed from the heel to the big toe. Remove your shoes and try this for yourself.
16 Take a number of steps in your bare feet. Describe what occurs from the time your left heel hits the ground until your right heel hits the ground. Referring to Figures 12.16 and 12.17 may help you with your description.
17 Summarise the main features in the human skeleton that are adaptations for an upright stance and for walking bipedally with a striding gait.

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CHAPTER 12 SUMMARY

- Humans are primates, and are in the same family as chimpanzees, bonobos, gorillas and orangutans. This family is called Hominidae, and the species are referred to as hominids.
- Primates have unspecialised bodies and limbs, five fingers and toes with nails instead of claws and an opposable first digit, forward-facing eyes, poor sense of smell, and a large and complex brain.
- Humans have a recent common ancestor with chimpanzees and gorillas, and a slightly more distant common ancestor with orangutans.
- Hominids share common features, including larger, more complex brains, five cusps on the molar teeth of the lower jaw, freely rotating arms, a wide, shallow chest, no external tail, an appendix and are active during the day.
- The size of the brain, especially the cerebral cortex, has increased during evolution. Humans have larger brains with more convolutions when compared to the other apes. This has allowed more advanced behaviours, such as tool making.
- As brains are not fossilised, the size of the brain is determined by the cranial capacity – the volume inside the cranium.
- As primates have evolved, the degree of opposability of the thumb and big toe (with the exception of humans) has increased. This has led to a precision grip in addition to the power grip.
- During evolution, there has been a change from quadrupedalism to bipedalism, with humans walking on two legs with a striding gait. Adaptations that allow bipedalism are:
  - central foramen magnum
  - S-shaped curve (double curve) of the spine
  - flatter facial profile with reduced protrusion of the jaw
  - a broad, shorter pelvis
  - femurs forming a carrying angle with the vertical line
  - knee joints that are larger on the outside and contain ligaments that resist the force acting on the knee
  - a tibia that is larger than the fibula
  - big toes that are non-opposable
  - both a longitudinal and a transverse arch in the feet
  - legs that are longer than the arms, lowering the centre of gravity
  - muscle tone to support the body.
- Bipedalism provided a number of advantages that led to a greater chance of survival during natural selection and, therefore, to its being passed on to future generations.
- A striding gait occurs when the hips and knees can be fully extended. Hominins (humans) are the only species that are able to do this.
- In a striding gait the weight is transferred from the heel along the outside to the ball of the foot, and then across to the big toe. The trunk rotates, and this is balanced by the swinging of the arms. At the same time, the carrying angle of the femur keeps the weight close to the central axis.
- Through evolution of hominids, the teeth and jaw have changed so that humans do not have a diastema, and do have smaller canines and incisors and a parabolic dental arcade. There has also been a reduction in prognathism and the size of the brow ridge.

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CHAPTER 12 GLOSSARY

Acetabulum The socket of the pelvis in which the head of the thigh bone fits

Adaptation A particular structure, physiological process or form of behaviour that makes an organism better able to survive and reproduce in a particular environment

Bipedalism Walking on two legs

Brow ridge A ridge of bone above the eye sockets of the skull

Carrying angle The arrangement of the thigh bones to form an angle to the vertical

Cerebral cortex The outer layer of the cerebrum, made up of grey matter

Cerebrum The largest part of the brain; made up of left and right hemispheres

Convolution An upward fold of the cerebral cortex of the brain; also called gyrus

Cranial capacity The volume of that part of the skull that is occupied by the brain

Cranium The part of the skull that contains the brain

Dental arcade The shape of the pattern made by the teeth as they are set in the jaw

Dental formula A formula that gives the number of each type of tooth in one-quarter of the jaw

Diastema A gap in a row of teeth; usually refers to a gap next to the canine teeth in primates, with canine teeth that are much longer than the other teeth

Diurnal Being active during the day

Endocast An impression of the inside of the brain case, either artificial or natural, made of rock or some other solid material

Femur The thigh bone

Foramen magnum The opening beneath the cranium through which the spinal cord passes

Frontal lobe One of the five lobes of each cerebral hemisphere

Hominid A member of the family Hominidae; includes humans and the other great apes (chimpanzees, gorillas, orangutans and bonobos)

Hominin A member of the tribe Hominini; humans, both past and present

Longitudinal arch The arch of the bones of the foot, running from front to back

Lumbar Describes the lower region of the spinal column; lumbar vertebrae support the lower back

Metatarsals The bones of the foot between the toes and the ankle

Muscle tone The partial contraction of skeletal muscles

Opposability The ability to use the thumb to touch the tips of each of the other digits on the hand

Pentadactyl Describes a limb with five fingers or toes

Power grip Force applied by the fingers and thumb towards the palm to transmit force to an object

Precision grip The grasping of an object between thumb tip and fingertip, as in holding a pencil when writing

Prehensile Grasping; refers to the digits of a hand or a foot that can grasp an object

Prognathism Having a protruding jaw

Quadrupedalism Walking on four legs

Striding gait A way of walking in which the hip and knee are fully extended

Transverse arch The arch of the bones of the foot, running from side to side

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CHAPTER 12 REVIEW QUESTIONS

Recall

1 To which of the primate families do humans belong? Who shares this family with us?

2 Describe the evolutionary trend evident in primates concerning the mobility of the thumb and the other digits.

3 a List the components of the skeleton that allow humans to adopt an erect posture.

b How do these components differ from the corresponding ones in a quadrupedal animal?

c What are the advantages and the disadvantages of an erect stance and bipedal locomotion?

4 Describe carrying angle, and compare the carrying angle of an ape with that of a human.

5 How does the wide pelvis and carrying angle of the femur enable humans to walk without the body swaying from side to side?

6 What is an endocast? What can it tell us about the size and shape of the brain?

7 Describe the major anatomical and functional developments that have occurred in hominid brains over the past four million years or so.

8 Describe the change in the shape of the face of hominids over the past four million years or so.

Explain

9 Explain how muscle tone helps to support the body against the force of gravity.

10 When we walk, our arms move in a coordinated way. Explain how arm movement helps stabilise the body of a human while walking.

11 Human dentition is said to be unique.

a List the differences between the teeth of a human and those of an ape, such as a gorilla.

b How has the dental arcade changed in hominins compared with that of an ape?

Apply

12 Chimpanzees have been observed using a range of simple tools, mainly associated with feeding. Describe the structural characteristics of chimpanzees that enable them to make and use tools.

13 For humans to be able to stand upright, a number of adaptations have taken place. Changes have occurred to the skull, vertebral column, pelvis, legs and feet. Describe how each of these has contributed – and how they have interacted – to enable humans to adopt an erect stance.

14 If you have seen chimpanzees or gorillas walking bipedally, you will have noticed that they sway from side to side as they walk. Explain why they cannot stride as humans do.

15 What assumptions are made when scientists infer the degree of intelligence from the cranial capacity of a skull?

16 The human canine tooth is much smaller than that of the other hominids, especially in the males of the species. Describe the evolutionary processes that would have taken place in hominins to produce the current size of that tooth in humans today.

17 Briefly describe how the environment could have contributed to the first hominins evolving the free striding gait. How would this gait have increased the chance of survival in that environment?

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Extend

18 As a result of various conditions, the normal curves of the vertebral column may become exaggerated. Use references to describe the conditions known as scoliosis, kyphosis and lordosis.

19 The term 'hominid' used to have the same meaning that 'hominin' now has. 'Hominid' was used to refer to the various members of the human family tree. Scientists who study human origins have changed the classification scheme by introducing a new level, the tribe. 'Hominid' is now defined in a much broader way so that it refers to all great apes and their ancestors. 'Hominin' refers only to present-day humans and our extinct ancestors. Why would scientists make changes to the classification scheme for apes and humans? Suggest as many reasons as you can.

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13 HOMININ EVOLUTION

UNIT 4 CONTENT

SCIENCE INQUIRY SKILLS

» select, use and/or construct appropriate representations, including phylogenetic trees, to communicate conceptual understanding, solve problems and make predictions

SCIENCE UNDERSTANDING

Hominid evolutionary trends

» determining relatedness and possible evolutionary pathways for hominins uses evidence from comparisons of modern humans and the great apes with fossils of:
  – *Australopithecus afarensis*
  – *Australopithecus africanus*
  – *Paranthropus robustus*
  – *Homo habilis*
  – *Homo erectus*
  – *Homo neanderthalensis*
  – *Homo sapiens*

» tool use is seen in a number of hominin species and the study of these tools provides important insight into the evolution of the human cognitive abilities and lifestyles. Trends are seen in the changes in manufacturing techniques and the materials used in the tool cultures of:
  – *Homo habilis*
  – *Homo erectus*
  – *Homo neanderthalensis*
  – *Homo sapiens*

Source: School Curriculum and Standards Authority, Government of Western Australia

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The apes, including humans, have the same basic characteristics and are classified in the family Hominidae. However, humans differ from other apes in their appearance and structure. Each animal species has developed adaptations that help it to survive and reproduce in its particular environment. Humans are no different, and we have developed features that set us apart from the other primates. As such, humans are classified as hominins; they belong to the tribe Hominini.

Hominins differ from other apes in their appearance, structure and behaviour. Most noticeably, hominins are relatively hairless compared with apes, and the structure of their upper and lower limbs allows for a fully bipedal way of walking. Humans stand and walk with an erect posture and a striding gait that is unique. It is not found anywhere else in the animal kingdom.

Hominins also have greater development of the brain, changes in the size and shape of the teeth, development of speech and sexual characteristics, all of which separate them from the other hominids.

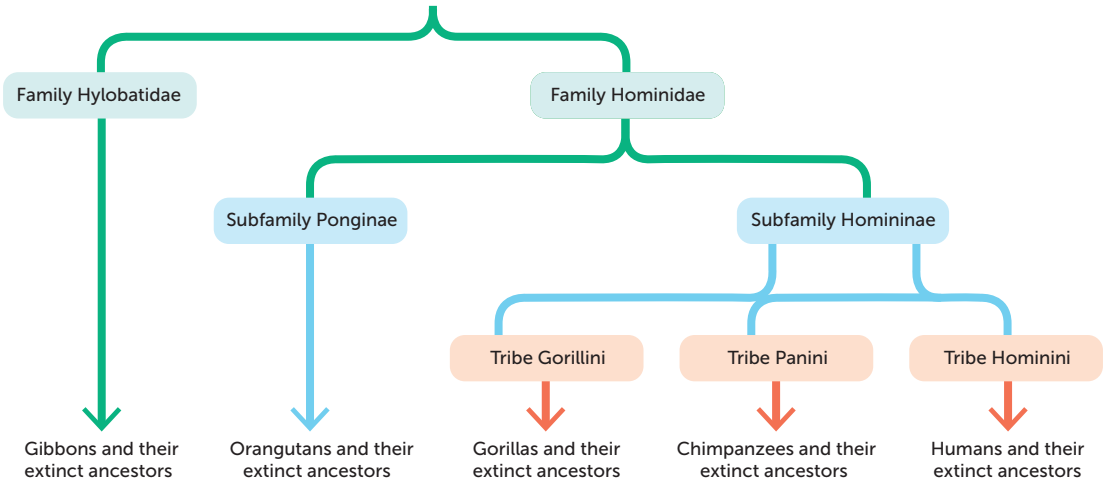

FIGURE 13.1 Classification of apes, including humans

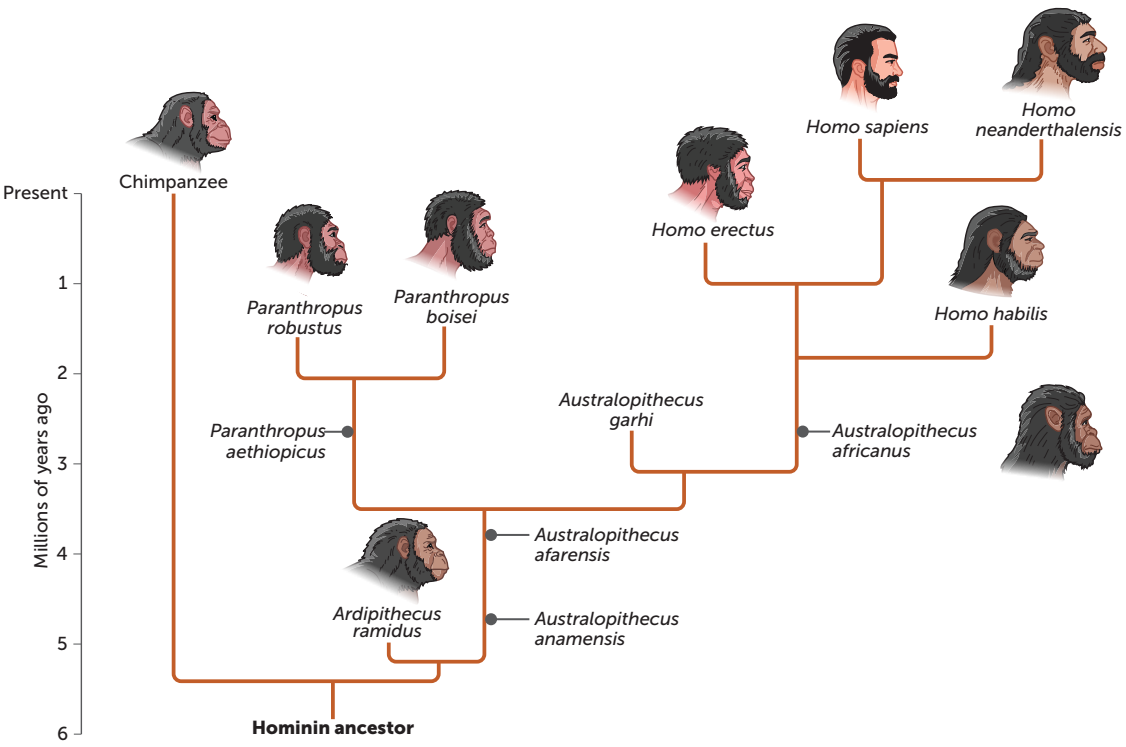

FIGURE 13.2 Illustration reflecting the evolution of hominins

Hominid and hominin
This website discusses the use of the terms 'hominid' and 'hominin'.

An interactive timeline
This website has an interactive timeline with detailed information about hominin evolution.

Throughout this chapter, we will be referring to the extinct ancestors of present-day humans. All hominids share a common ancestor, an ape-like creature. From that ancestral ape the first hominins evolved.

The evolutionary trends described for primates in Chapter 12 continue in the hominins. However, hominins are set apart from the other hominids by some very special adaptations that give them a unique position in the animal kingdom.

In this chapter, we will look at the characteristics of a number of species of hominins that show the evolutionary changes leading to present-day humans.

13.1 EVOLUTIONARY TRENDS IN HOMININS

During human evolution there is a general trend of increasing cranial capacity and skull size along with reduced prognathism. *Homo sapiens* also have a reduced brow ridge compared to earlier species.

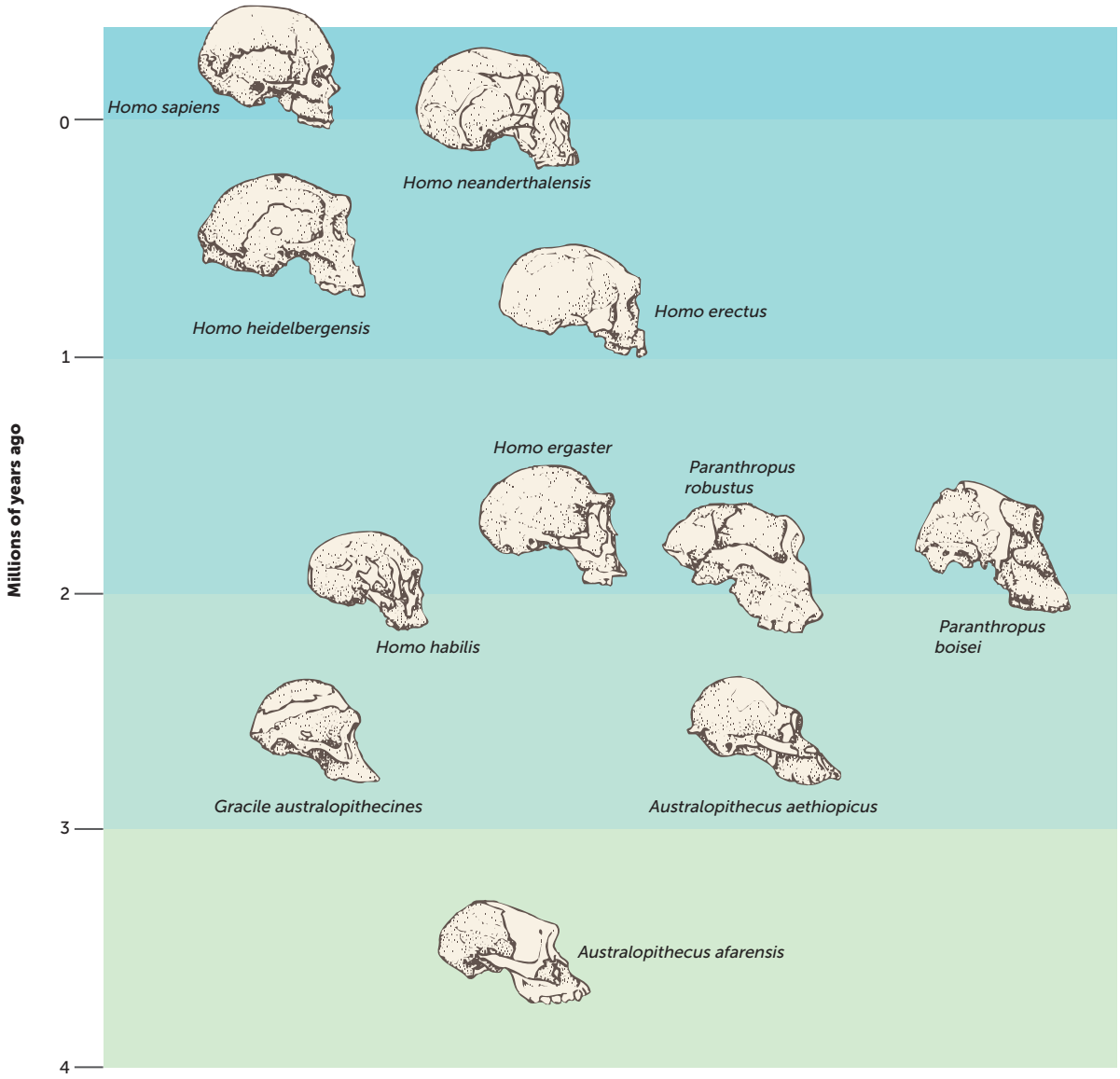

Figure 13.3 Fossil hominin skulls from different time periods. Note the increased cranial capacity and reduced prognathism from the early australopithecines to modern humans.

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Relative size of the cerebral cortex

Unlike bipedalism, which was well established in early hominins, the gradual increase in the size of the cranium to house a larger and more complex brain is an evolutionary trend in hominins. **Endocasts** have been used to calculate the **cranial capacity** of fossilised skulls. This has enabled scientists to infer that early hominins such as *Australopithecus afarensis* had a cranium that was much closer in size to that of modern apes than modern humans.

Subsequent fossil evidence confirmed a gradual increase in cranial capacity as the hominin species evolved towards modern humans. The average brain size of the first australopithecine fossils found placed them within the range of modern gorillas. However, the body weight of these fossil australopithecines was probably only a third that of the gorilla, so their *relative* brain size lay somewhere between that of chimpanzees and modern humans.

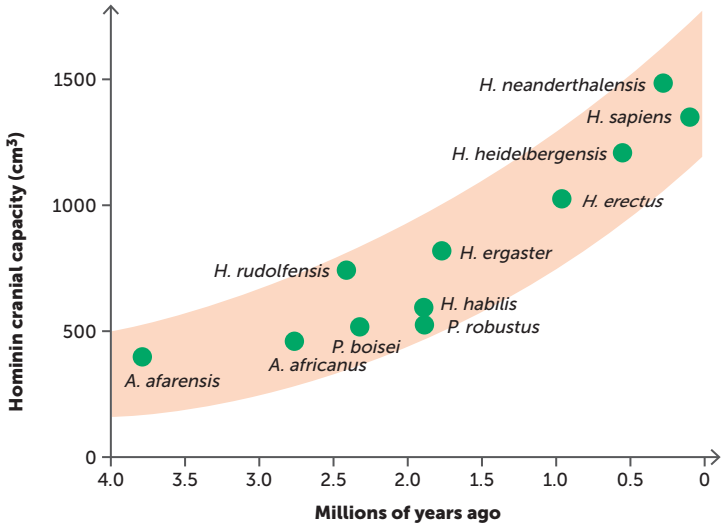

FIGURE 13.4 Graph demonstrating the gradual increase in cranial capacity of hominins over time. Average cranial capacity is shown for each species.

The endocasts of australopithecines also indicate that their foramen magnum was more forward than it is in the apes, and the skull more rounded at the back.

Sometimes only fragments of fossil skulls are found. Without an endocast, determining cranial capacity is very difficult, and even experts vary in their estimates. For example, when the first specimen of *Homo habilis* was discovered in Olduvai Gorge, Tanzania, in 1960, three different anthropologists gave three varying estimates for the cranial capacity: 590 cubic centimetres (cm^3), 647 cm^3 and 710 cm^3. Such a range of figures from an examination of the same material shows that estimates of cranial capacity must be treated with caution. The averages listed in Table 13.1 must be considered approximations at best.

TABLE 13.1 Hominin cranial capacities

HOMININ	CRANIAL CAPACITY (cm^3) (ESTIMATE OF BRAIN SIZE)
Australopithecus afarensis	430
Australopithecus africanus	457
Australopithecus garhi	450
*Paranthropus boisei**	491
*Paranthropus robustus**	542
Homo habilis	590
Homo rudolfensis	774
Homo ergaster	800
Homo erectus	1004
Homo heidelbergensis	1226
Homo neanderthalensis	1485
Homo sapiens	1350

Note: *Many classification schemes include the genus *Paranthropus* in the genus *Australopithecus*.

Activity 13.1
Investigating cranial capacity and phylogenetic trees

Fossil endocasts reveal more than just an increase in cranial capacity. A gradual increase in the number of convolutions and the size of the frontal lobe is also evident. These trends can be seen in *Homo erectus* fossils. Over the period of time that this species lived on Earth, the cranial capacity of *H. erectus* increased from about 750 cm^3 to 1250 cm^3. As the brain case expanded, the face tended to become flatter and a noticeable forehead began to develop in the later members of the species. This was probably due to an expanding frontal lobe.

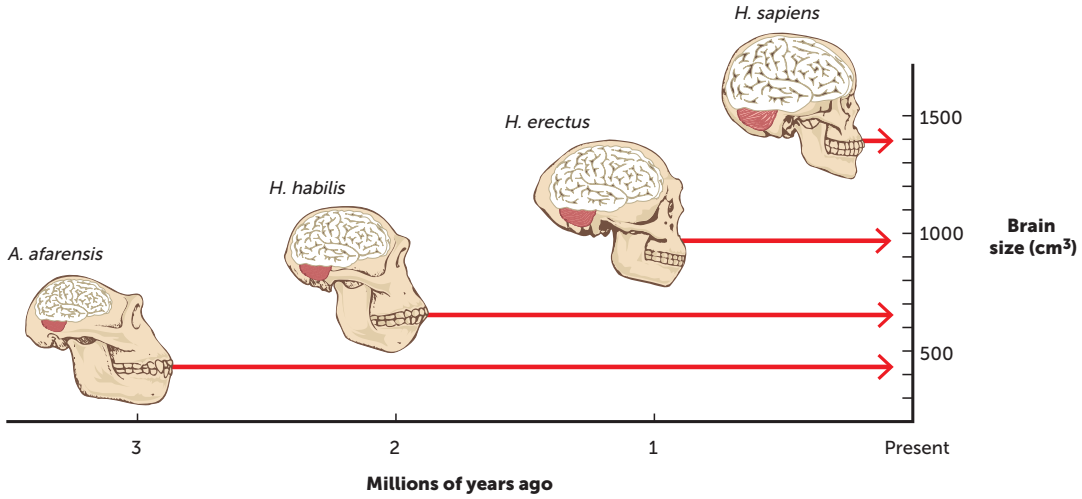

FIGURE 13.5 The increase in brain size in four hominin species over time. Note the marked expansion of the frontal region.

Increasing brain size
This website provides information on the increase in brain size in hominins.

Prognathism and dentition

The change in the dental arcade of hominins is another discernible evolutionary trend. Early hominins, such as *Australopithecus afarensis*, had a lower jaw and face that was more like that of other apes. The teeth were large and there was a distinct gap between the canines and the incisors, with the rows of teeth parallel rather than curved. However, by the time of *Homo habilis*, the molar and premolar teeth had become smaller and narrower, but the canines were still prominent, as can be seen in the fossil in Figure 13.6.

The trend towards smaller molars and a decrease in the robustness of the teeth continued in *Homo erectus* and is noticeable in modern humans. Humans that lived about 100 000 years ago had teeth that were about 10% larger than humans of today. Modern humans also appear to be gradually losing their wisdom teeth (the third molar), with an increasing number of people having no wisdom teeth at all.

Figure 13.7 shows the gradual enlargement of the cranial portion of the skull to accommodate the increasing size of the frontal region of the brain. This led to a more distinct forehead and to a reduction in prognathism and in the size of the brow ridge.

FIGURE 13.6 Fossil skull of *Homo habilis*

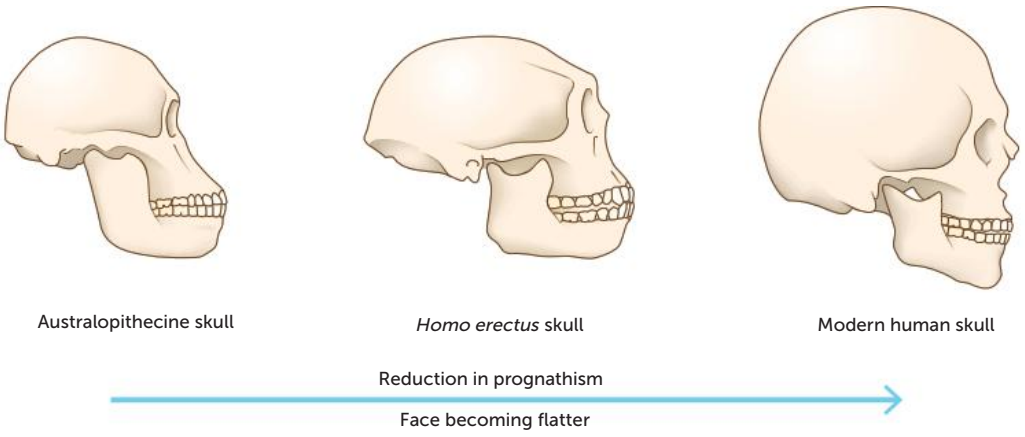

FIGURE 13.7 The evolutionary trend towards a flatter face

Reduction of prognathism
This website provides an excellent series of images illustrating the reduction in prognathism in hominins over time.

Activity 13.2
Investigating hominid skulls

Table 13.2 summarises the anatomical trends in human evolution.

TABLE 13.2 Anatomical trends in hominin evolution

ANATOMICAL FEATURE	CHARACTERISTICS MORE APE-LIKE (CONSIDERED TO BE MORE PRIMITIVE)	CHARACTERISTICS MORE HUMAN-LIKE (CONSIDERED TO BE MORE MODERN)
Skull	Thicker bones forming cranium Face large compared to cranial size Smaller cranial capacity Heavier brow ridges No forehead or sloping forehead Lower cranium Less prominent cheek bones Possible saggital crest on top of skull Foramen magnum towards back of skull (post-central)	Thinner bones forming cranium Face small compared to cranial size Larger cranial capacity Brow ridges reduced or absent Increasingly larger and more vertical forehead More dome-shaped cranium More prominent cheek bones No crest on top of skull Foramen magnum under centre of skull
Mandible and teeth	More prognathic jaw Larger jaw Heavier, thicker mandible No chin Larger teeth, especially molars Diastema present Canine teeth more prominent Difference between size of incisors and molars	Flatter face Smaller jaw More slender, thinner mandible Increasingly definite chin Smaller teeth No diastema Canine teeth less prominent More even teeth/\little difference in size of incisors and molars
Torso	Narrower pelvis Back (lumbar) vertebrae less wedge-shaped Wide, barrel-shaped ribcage	Broader pelvis Lumbar vertebrae more wedge-shaped Smaller ribcage
Upper limbs	Shorter thumb that is less mobile Fingers longer and more curved	Longer thumb with increased opposability Fingers straighter and shorter
Lower limbs	Femurs more parallel Arms longer than legs	Femurs sloping inwards towards the knee Arms shorter than legs

Key concept

Evolutionary changes in hominins include an increased cranial capacity and convolutions, as well as a decreased size of the teeth, diastema and prognathism.

Questions 13.1

RECALL KNOWLEDGE

1. Which tribe do humans belong to, and what other species are also in this tribe?
2. Explain the difference between brain size and cranial capacity.
3. State the trend in cranial capacity and convolutions of hominins.
4. Define 'endocast' and describe how it is used to infer the shape of the brain.
5. State the cranial capacity of:
   - a *Australopithecus afarensis*
   - b *Australopithecus africanus*
   - c *Paranthropus robustus*
   - d *Homo habilis*
   - e *Homo erectus*
   - f *Homo neanderthalensis*
   - g *Homo sapiens*

APPLY KNOWLEDGE

6. Explain why the cranial capacity of a fossilised skull is used to infer brain size.
7. Explain the relationship between changes in the size of teeth and prognathism.

13.2 COMPARISON OF HOMININ SPECIES

Genus *Australopithecus*

Fossil evidence of australopithecines

The first australopithecine fossil was found in southern Africa in the early 1920s. Like many early fossil discoveries, it was a chance event. Raymond Dart, a young Australian anatomist, had his attention drawn to fossil baboon skulls being found in limeworks at Taung, north-west of Kimberley in South Africa. Dart asked the manager of the limeworks to send him any interesting fossils, which he did, sending a box full of limestone pieces containing bones. On clearing away the limestone, Dart was surprised to find the whole face, jaws and teeth of what appeared to be an ape. However, it was like no other ape: although it was a juvenile, Dart realised that the face was not as protruding as that of an ape, and the teeth, especially the first molars, were more like those of humans. The skull was more rounded, and there was no brow ridge (Figure 13.8). Dart's account of his discovery was published in *Nature early* in 1925. In his article, Dart suggested that the skull should be named *Australopithecus africanus*, 'the southern ape of Africa', and that it be put in a new family midway between apes and humans.

Alamy Stock Photo/The Natural history Museum

FIGURE 13.8 The Taung skull: Side view of a cast of the original fossil material

The Laetoli footprints are evidence that early hominins existed over 3 million years ago. Although there have been a number of interpretations of these footprints, with different numbers and sexes for the individuals who made them, most scientists agree that they were made by *Australopithecus afarensis* (a separate species of australopithecines) 3.56 million years ago. Features of the footprints that indicate a bipedal form of locomotion include a deep impression showing the heel hitting the ground first, the lateral transmission of weight from the heel to the ball of the foot, a well-developed longitudinal arch, a big toe that was parallel to the other digits, and a deep impression where the toe pushed the foot forward for the next stride.

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Another important discovery was the fossil remains known as 'Lucy', which were found in the Hadar region of Ethiopia (see Figure 11.1 in Chapter 11), along with several hundred fossil fragments. The fragments are thought to be of individuals who lived and died near a now-vanished lake between 3 and 3.6 million years ago. 'Lucy' is a female skeleton that was 40% complete. 'Lucy' has been classified as *Australopithecus afarensis* based on evidence gained from the dental arcades, the size of the canines and the prominence of the cusps on the cheek teeth.

Science Photo Library/John Reader

FIGURE 13.9 The Laetoli footprints – footprints made in volcanic ash 3.56 million years ago. More than 3 million years ago, the ancestors of modern humans were walking in very much the same way that we do today.

Features of australopithecines

From the fossil evidence so far accumulated, it has been possible to construct a clear picture of the physical features of *Australopithecus*. Many of these resemble the features of later hominins. The teeth are typically those of a hominin: the canines are short and non-projecting, resembling the incisors, in sharp contrast to those of other apes. Together the incisors and canines make a row of cutting teeth, and there is no gap between them and the following premolars. The teeth are in the parabolic shape distinctive of the hominids.

Finding 'Lucy'
This website provides an interesting video on the discovery of this fossil.

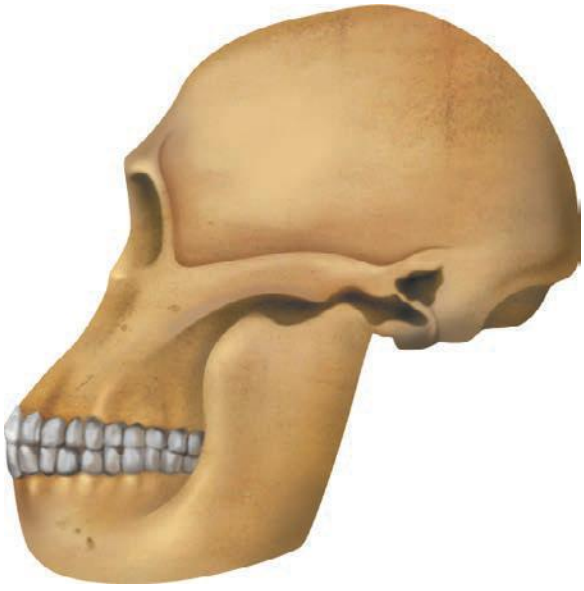

FIGURE 13.10 Reconstruction of an australopithecine skull

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The facial profile of the australopithecines has a low forehead, and a more projecting upper and lower jaw than more modern hominin profiles. The average brain size is around 480 cm^3, which is within the range of that of modern gorillas. However, the australopithecine's body weight was probably only a third that of the gorilla, and so their relative brain size lies somewhere between that of chimpanzees and modern humans.

Considering the evidence from fossil bones and fossil footprints, it is safe to assume that these early hominins were truly bipedal, even though their gait would not have been quite the same as that of modern humans. The femur, pelvis and carrying angle in australopithecines are much more like those of a human than an ape, as Figure 13.11 indicates. The pelvic and foot bones are typically hominin, with the foot possessing a non-opposable, strongly built, robust big toe. Additionally, the foramen magnum was more forward than it is in the other apes, and the skull more rounded at the back. Finally, the vertebral column displays the typical hominin 'S'-shaped curvature, which, together with the central position of the foramen magnum, indicates an erect stance.

Bones of the hand suggest that the thumb was shorter and less mobile than that of modern humans, and the fingers more heavily built, features indicating that the hand was better adapted for the power grip than the precision grip. This may indicate an arboreal lifestyle.

Australopithecus species

While there are many similarities between all australopithecines, there are also some variations, indicating the evolutionary changes. These are summarised in Table 13.3.

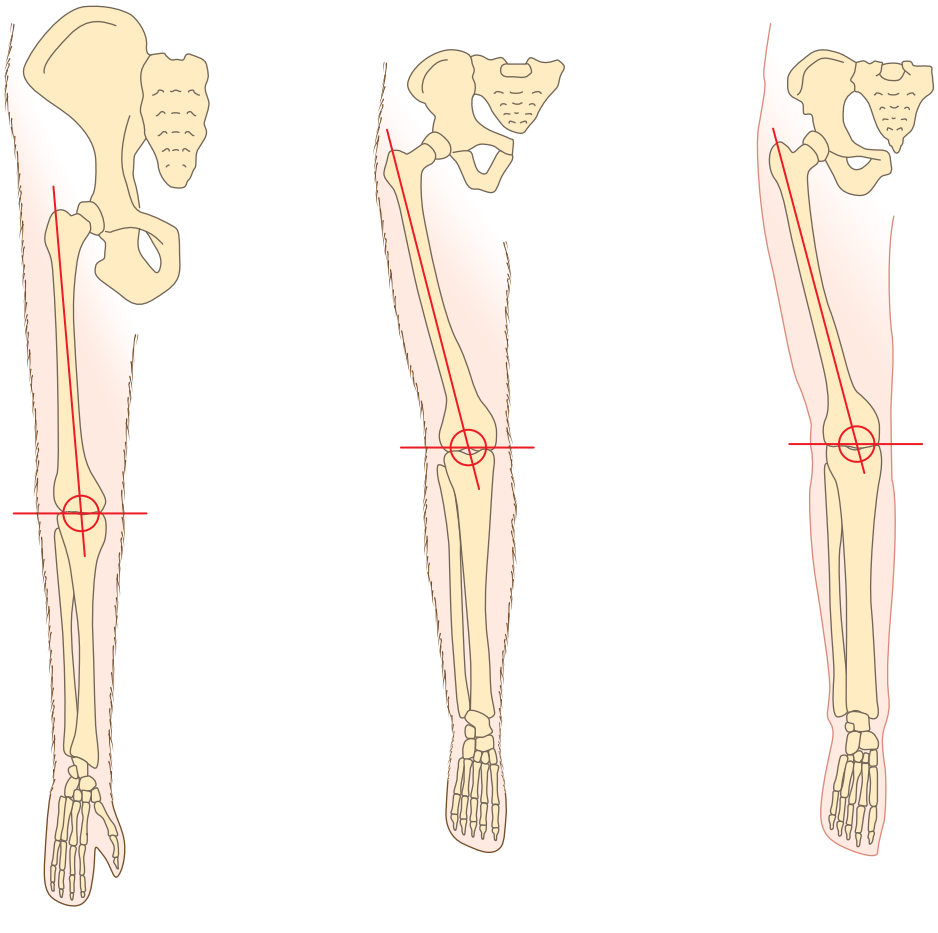


FIGURE 13.11
Australopithecines, like modern humans, had the femur angled so that the foot was under the centre of gravity, allowing bipedal locomotion with the striding gait. The femur of other apes is not angled in this way, so they sway from side to side when walking erect.

TABLE 13.3 A comparison of *Australopithecus afarensis* and *Australopithecus africanus*

	Australopithecus afarensis	*Australopithecus africanus*
Time of existence	3.9 and 2.8 million years ago	3.2 to 2 million years ago
Location	East Africa	Southern Africa
Height	Female: 105–110 cm Males: 150 cm	Female: 110 cm Males: 135 cm
Brain	430 cm^3	480 cm^3
Skull	Low, sloping forehead Prominent brow ridges Short sagittal crest in males	Slightly arched forehead Smaller brow ridge
Teeth and jaw	Prognathic jaw Small canine teeth (but larger than *A. africanus*) Diastema present	Prognathic jaw Shorter and smaller incisors and canines Large molar and premolars No diastema
Limbs	Big toe not opposable Long arms, although shorter than the legs Long curved fingers and toes Features for bipedalism	Big toe not opposable Long arms, although shorter than the legs Some curvature of the finger and toe bones Features for bipedalism
Pelvis	Short and wide pelvis	Short and wide pelvis, less rounded than in modern humans

Australopithecus afarensis
This website has more information about *Australopithecus afarensis*.

Australopithecus africanus
This website has more information about *Australopithecus africanus*.

Paranthropus robustus

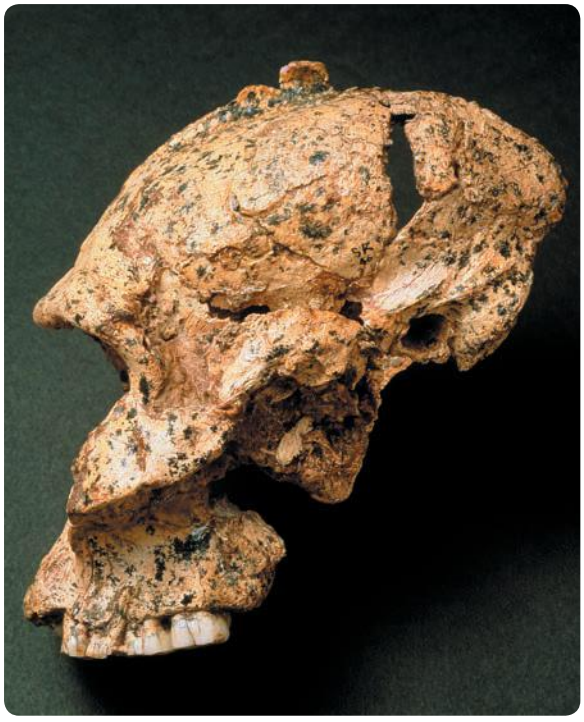
Robert Broom discovered a fossil jaw fragment and molar in 1938 with large molars and a strongly built jaw. This was different from the features of the human species known at that point. This led Broom to believe it was evidence of another species, *Paranthropus robustus*.

Individuals in the *Paranthropus robustus* existed in South Africa about 1.8–1.2 million years ago. The species has been extinct for more that 1 million years. It is thought that they are not an ancestor of modern humans, but instead formed part of an evolutionary branch with no descendants. This can be seen in the phylogenetic tree in Figure 13.2.

Paranthropus robustus used to be classified as a robust australopithecine based on the robust jaw and skull. However, it is now considered to be a separate genus.

Given that they were originally classified as australopithecines, it makes sense that *Paranthropus robustus* share many of the same characteristics as australopithecines. Some of the key features of *P. robustus* are:

- females' height of approximately 1 m and males' height of approximately 1.2 m
- cranial capacity of 520 cm^3
- large sagittal crest for attachment of strong chewing muscles
- very large molars and premolars, with small incisors and canines by comparison
- prognathism, although less than australopithecines
- wide, dish-shaped face with large zygomatic arches
- heavy brow ridges
- structures for bipedalism.



Science Photo Library/John Reader

FIGURE 13.12 A fossil of *Paranthropus robustus* found in 1936

***Paranthropus* genus**
This website has more information about *Paranthropus* genus.

Homo habilis

In 1964, Dr Louis Leakey published an account of a new species of *Homo* found at Olduvai Gorge in East Africa. Together with two colleagues, Professor Phillip Tobias and Dr John Napier, he had found a jaw, two cranial fragments, and several post-cranial remains dating back to 1.75 million years BP. They gave the new species the name *Homo habilis*, or 'handy human', to indicate that it was adept at tool making. Usually the announcement of something 'new' in science causes other authorities in the field to question the interpretations of the discoveries. The case of *H. habilis* was no different. Many authorities considered it to be nothing more than an advanced australopithecine, or an East African variant of *Australopithecus africanus*. However, *H. habilis* had a larger brain and smaller teeth than the australopithecines, suggesting that their diet included meat. They were taller than the gracile forms and stood more erect. At the time of its discovery it was thought to be the earliest tool user.

Homo habilis lived between 2.3 and 1.5 million years ago in eastern and southern Africa. Individuals show features between apes and humans. These include:

- females' height of 110 cm and males' height of 130 cm
- brain size of 610 cm^3
- rounder skull
- small brow ridge
- central foramen magnum
- moderate prognathism
- teeth arranged in a rounder arc
- relatively short legs and long arms
- slightly curved finger bones, indicating a strong power grip
- able to form a precision grip.

Science Photo Library/Natural History Museum, London

FIGURE 13.13 Cast of a *Homo habilis* skull discovered in 1973 in Kenya

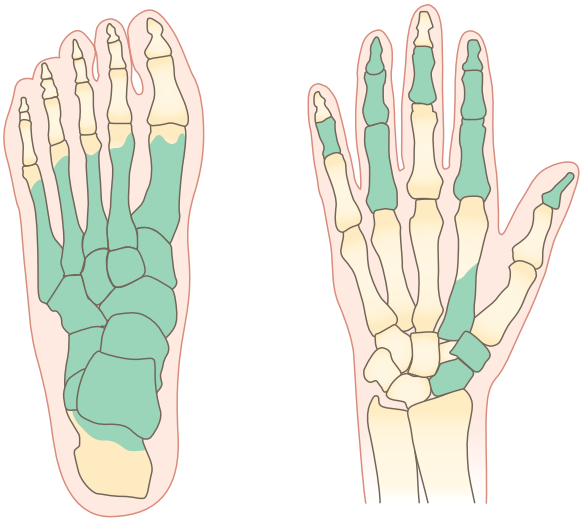


FIGURE 13.14 Hand and foot fossil bones (shown shaded) of *Homo habilis;* they resemble those of modern hominins. There is evidence of heavy musculature, indicating a powerful grip

Homo habilis
This website provides more information about, and images of, *Homo habilis*.

Homo erectus

Homo erectus were the first humans to show modern, human-like bodies, indicating a life on the ground rather than in the trees.

In 1927, Dr Davidson Black announced that he had found a new species, which he called *Sinanthropus pekinensis*, or Chinese human of Peking (now known as Beijing). This is why the fossil is known as 'Peking Man'.

Alamy Stock Photo/ImageBROKER

FIGURE 13.15 Reconstruction of a *Homo erectus* skull based on fossil remains found at Zhoukoudian Cave in China

Peking Man
This website suggests that 'Peking Man' may have been more sophisticated than was once thought.

Black's announcement came after the study of teeth found in a limestone cave near Zhoukoudian, south of Beijing. Two years later the first skull was found; in the years to follow, four more skulls, plus skull fragments, lower jaws and teeth, were discovered. These fossils are now included in the species *Homo erectus*, and they are still some of the best examples ever found. Unfortunately, during World War II the original fossils were lost, but good plaster casts had been made of them and each had been extensively described in the scientific literature, so the material may still be studied today.

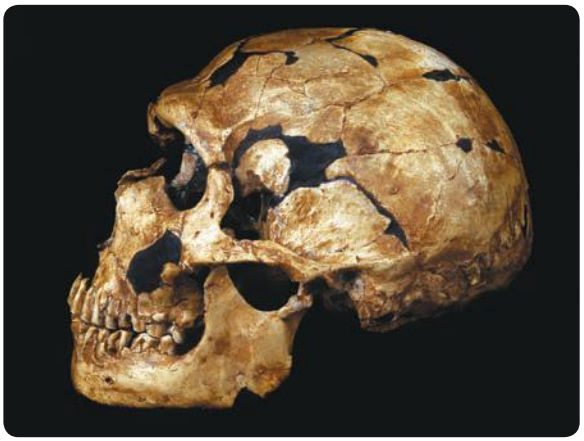
When compared to fossils of earlier human species, the brain of the Beijing specimens was considerably larger, with an average size of 1075 cm^3, and some aspects of the skull showed more modern features. The curve of the dental arcade was shorter and more rounded in front. The jaw was shorter and more compact, and suggested that a chin was beginning to form. Finally, the teeth were very modern and indicated a diet much like that of humans today. Evidence of the use of fire was also found in the cave, together with the remains of small, quartz, flake-like tools and animal bones.

Some key features of *Homo erectus* are:

- varied height, ranging from 145 cm to 185 cm
- short, stocky body with thicker bones, suggesting a demanding lifestyle
- average cranial capacity of 1050 cm^3
- low, sloping forehead
- defined brow ridges
- large, thick jaw without a chin
- reduced size of molars.

Homo neanderthalensis

The first recognised fossils of Neanderthal people were found in 1856 in a cave in the Neander Valley, near Düsseldorf, Germany. Since then a great many more fossils of this type have been found throughout Europe, Asia and northern Africa. Interpretations of data from the fossils have varied in the past, but fossils found in the 1990s suggest that the Neanderthals were only a side-branch along the pathway to modern humans. This was confirmed when, in 1997, molecular biologists extracted some DNA from a Neanderthal fossil and compared it with that of modern humans. They concluded that the Neanderthals were a distinct biological species, *Homo neanderthalensis*. They existed in Europe during the last of the ice ages and were adapted to that particularly harsh type of environment. At some time in the past, the lineage diverged, with one branch leading to Neanderthals and another to modern humans. Exactly when, and how, this split took place we do not know, but there is considerable evidence that for a time Neanderthals and *Homo sapiens* lived together in Europe.



Science Photo Library/Javier Trueba/MSF

FIGURE 13.16 Neanderthal skull showing the 'bun' shape at the back

Neanderthals, while clearly human, had many features that evolved due to a cold, harsh climate. They had big faces, low but large skulls, and heavy brow ridges. The brain was slightly larger than the average for humans today, and its shape was different. The back of the skull was drawn out in a 'bun' shape, the lower jaw lacked a definite chin, and the cheeks were swept back to give a streamlined appearance. These features can be seen in Figure 13.17, where the skull of *Homo neanderthalensis* is compared with those of *H. erectus* and *H. sapiens*. Note how it appears to have some features of both. The robust nature of the Neanderthal skull echoes the physical appearance of *H. erectus*, while the much larger brain of the Neanderthals is considered a modern feature. Peculiar to the Neanderthals is the forward thrust to the face, or prognathism, accentuated by the way the nasal bone projects forward. It is believed that the Neanderthal nose projected more than the modern human nose and was much wider. This larger, wider nose is thought to have been an adaptation for life in seasonally cold and dry environments.

Homo neanderthalensis
This website has more information about *Homo neanderthalensis*.

Neanderthal brains: Bigger, not necessarily better
This article compares the brains of Neanderthals and modern humans.

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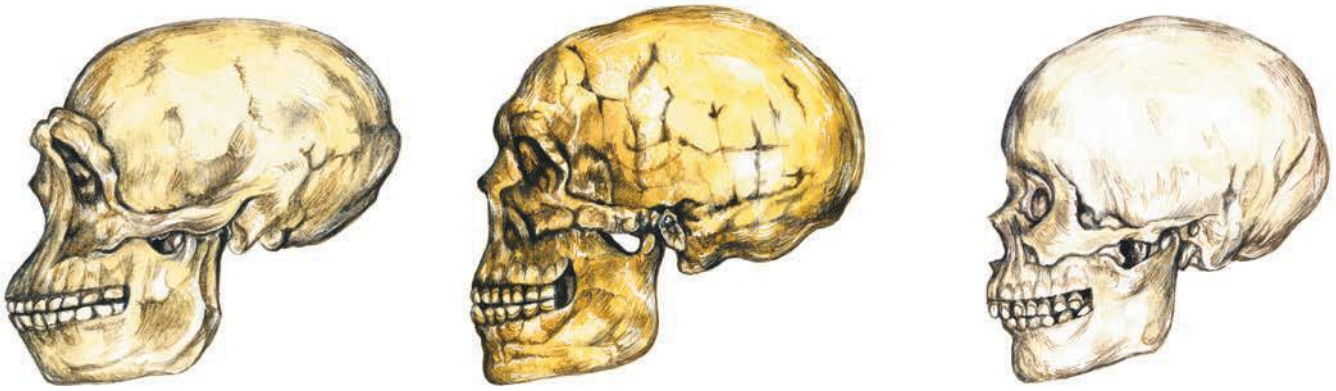


Science Photo Library/Christina Brodie, Visuals Unlimited

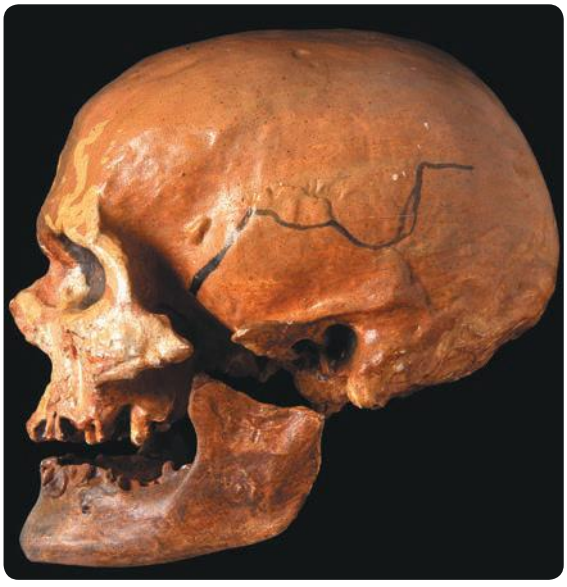
FIGURE 13.17 Skulls of *Homo erectus* (left), *Homo neanderthalensis* (centre) and a modern human (right)

The Neanderthals were short in stature, males being probably a little more than 1.5 metres in height, and females a little shorter. The limbs were short and heavily jointed with powerful muscles, so they would have appeared much more heavily built than modern humans. A barrel-shaped chest and thick neck muscles would have added to the rugged appearance. On top of this solid frame was a large skull containing a brain that was, on average, slightly larger than normal for modern humans, averaging 1485 cm^3, compared to 1350 cm^3 for modern humans. It has been suggested that the additional brain capacity was probably required for control of the extra muscles. Apart from these differences in physical characteristics, Neanderthals would have walked, run and used their hands in much the same way as modern humans. Table 13.4 summarises the key features of *Homo neanderthalensis*.

Homo sapiens

When the first fossils of modern humans were found in Europe, no one realised their significance or importance. It was not until 1868 that fossils of this type attracted the attention of scientists. In that year a number of skeletons were found at Cro-Magnon, under an overhanging cliff near the village of Les Eyzies in France. These fossils, of what are now called the **Cro-Magnon people**, were discovered by workmen constructing a railway. The site revealed the remains of more than five people, together with animal bones, seashells in the form of necklaces, and stone tools. The stone tools were similar to those that had been found at Aurignac, tools that had become known as Aurignacian. Later discoveries suggested that these fossils were part of a once-widespread population distributed throughout Europe from 40 000 to about 12 000 years ago. The best records of this habitation date from 25 000 years ago and occur in Spain, the French Pyrenees and the Dordogne Valley in France.

Cro-Magnon people were members of our own species, *Homo sapiens*, and they possessed features far more modern than those of Neanderthals. In particular, their skulls tended to be shorter from front to back, higher in the region of the top of the skull and rounder at the back. Besides these, other features included less prominent brow ridges, a reduction in the projection of the face, and a smaller jaw, as can be seen in Figure 13.18. They had large brains, around 1350 cm^3 on average, housed in skulls that were long from front to back. The face was relatively broad and short, with the orbits, or eye sockets, well separated. The teeth also tended to be smaller and a chin had developed.



Science Photo Library/Natural History Museum, London

FIGURE 13.18 Cro-Magnon skull

Human lineage
This website provides an interactive timeline showing our hominin ancestors.

Table 13.4 shows the key information about *Homo neanderthalensis* and *Homo sapiens*.

TABLE 13.4 A comparison of *Homo neanderthalensis* and *Homo sapiens*

	Homo neanderthalensis	*Homo sapiens*
Time of existence	Between 28 000 and 300 000 years ago	300 000 years ago to present
Location	Europe and the Middle East	Worldwide
Body type	Shorter, more robust and muscular than modern humans Wider shoulders	Short, slender trunks and long limbs
Height	Females: 156 cm Males: 168 cm	Females: 160 cm Males: 175 cm
Brain	1500 cm^3	1350 cm^3
Skull	Long and low brain case Occipital bun at the back of the skull Thick brow ridges Receding forehead, elongated skull Flared zygomatic arches Depression at back of skull for neck muscle attachment	Short base and high brain case
Teeth and jaw	Larger, more robust prognathic jaw Lacking a chin Larger teeth	Short jaw Bony chin Small teeth
Limbs	Thick limbs with large joints Shorter	Long legs compared with the arms Straight fingers and toes
Pelvis	Wider pelvis	
Ribcage	Barrel-shaped	Less barrel-shaped

Key concept

During the evolution of hominins, there has been a general increase in cranial capacity and height along with a decrease in prognathism and brow ridges.

Questions 13.2

RECALL KNOWLEDGE

1 Complete the following table for the species listed.

SPECIES	TIME OF EXISTENCE	HEIGHT	KEY PHYSICAL FEATURES
Australopithecus afarensis			
Australopithecus africanus			
Paranthropus robustus			
Homo habilis			
Homo erectus			
Homo neanderthalensis			
Homo sapiens			

2 Which species studied were the first hominin to show bodies similar to modern humans?

3 Describe features of fossils of australopithecines that would indicate bipedalism.

4 What is a key feature that will allow the identification of a *Homo neanderthalensis* skull?

WS
13.1 Human evolution

Activity 13.3
Investigating evidence for human evolution

Activity 13.4
Are humans unique?

5 Describe the fingers of both *Homo habilis* and *Homo erectus*. Use this to justify which of the two has a more common ancestor with modern humans.
6 Explain why the forehead of *Homo sapiens* is rounder and higher than earlier species.
7 Describe the features of the skull of a *Homo sapiens*.

APPLY KNOWLEDGE

8 Classify the hominin species of the skull to the right. State the features used in your classification.
9 Explain how the fossilised Laetoli footprints would have been produced.
10 Explain the relevance of a large sagittal crest in skulls of *Paranthropus robustus*.
11 Neanderthals lived in cold, harsh climates. Discuss two physical features that would have evolved in this environment.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13.3 CULTURAL EVOLUTION

Anthropologists, people who study human societies and their development, may define **culture** as anything that is learnt. Thus, activities such as making stone tools, hunting techniques, food preparation, using language and making art are all part of culture.

Just as the physical characteristics of hominins evolved over time, hominin culture has also evolved. Cultural development was an important means of overcoming some of the environmental challenges faced by early humans. This **cultural evolution** can be seen in the gradual improvement in tools, better methods of obtaining food, increased sophistication of language and a host of other changes culminating in the highly complex culture that we have today.

Tool use by australopithecines

The areas once occupied by australopithecines reveal the existence of **home bases**, from where hunters and foragers went out to search for food. No evidence of the use of fire by australopithecines has been found to date, but tool use does appear to have been common. A range of **pebble tools** have been found, including choppers, scrapers, flakes and chisels. These vary from about the size of a tennis ball (choppers) to that of a marble (scrapers and flakes), and are frequently referred to as **Oldowan tools**, after the site where they were first discovered. To use the scrapers effectively, the precision grip must have been employed. Tools of this type have been found at sites dating back 2.5 million years.


Alamy Stock Photo/The Natural History Museum

FIGURE 13.19 Oldowan choppers

This early tool making marked the start of a change in the way hominins interacted with their environment. They used items present in their surroundings, such as pebbles, sticks and plant material. However, there is no evidence suggesting that they changed these tools. These simple pebble tools enabled the australopithecines to exploit the resources in their environment more effectively and were the first stage in a succession of cultural changes still going on today.

Tool use by australopithecines enabled them to exploit a broader range of habitats, so they were eventually able to leave Africa and colonise other continents. Evidence suggests that the australopithecines began to disperse from Africa around 2 million years ago.

Tool use by *Homo habilis*

Homo habilis continued to use Oldowan tools. Some of these were sharpened or shaped by striking one stone with another. These would have been used for activities such as skinning animals, chopping up meat, breaking open bones, crushing plants and digging up edible plant roots.

Homo habilis lived in grasslands and were hunter-gatherers. Their diet would have been primarily plant material, with supplementary meat from either scavenging or hunting. The meat would have provided the complex fats needed for brain growth. This corresponds with the evidence of an increase in cranial capacity.

Typically, hunter-gatherers would have worked in groups, with specific members being responsible for different tasks. Those collecting food would have brought it back to the home base to share among the members. This indicates a social organisation in *Homo habilis*. Communication within the group would have been important, and thus pressure for development of a spoken language would have increased. There is some evidence that early *Homo* had a bulge in the speech-producing area of the brain, but the larynx may not have been capable of making complex sounds.

Evidence that early *Homo* was both a hunter and a scavenger of meat comes from animal bones found at fossil sites. A number of the bones show cut marks made by stone tools. With the naked eye it is difficult to distinguish between cut marks made by stone tools and those made by the teeth of a carnivore. However, examination under high magnification shows a clear distinction between the two. In Figure 13.20, notice how the tooth has left a broad, smooth groove on the bone, whereas the stone tool has made smaller, parallel grooves in the main cut. When interpreting the meat-eating behaviour of these early hominins, it is important to determine which cut marks were made first. Were they scavenging the remains of prey killed by carnivores, or were they consuming meat from animals they had killed and butchered? The bones recovered suggest that they were engaged in both activities. It is likely that *Homo habilis* were more scavengers and that as *Homo* evolved, hunting became more important.

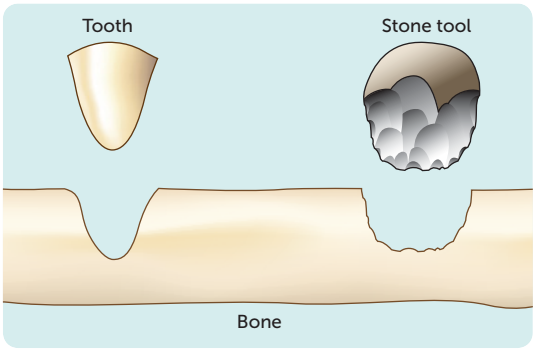

FIGURE 13.20 Difference in the marking patterns on bone produced by a tooth and by a stone tool

Tool use by *Homo erectus*

By the time of *Homo erectus*, the effect of the environment as a selective agent was diminishing. These hominins were now modifying the environment to suit their own purposes. The use of fire, the building of shelters and a range of sophisticated tools had enabled *H. erectus* to become more independent of the environment.

Science Photo Library/Pascal Goetgheluck

FIGURE 13.21
Acheulian hand axe

Tools manufactured by *H. erectus* were flaked around all of the edges, first in one direction and then in the other, until they formed roughly two-faced (bi-faced) lumps, approximately teardrop in shape. These tools were used as hand axes and are usually referred to as **Acheulian tools**, after the site at St Acheul in France where they were first discovered.

The discovery of a site on the Riviera in France in 1966 revealed much about the life of *H. erectus* in Europe 400 000 years ago. The site, called Terra Amata, contained 21 levels of habitation. Among important discoveries were the imprint of an adult foot, evidence of fire use and signs that *H. erectus* had constructed huts for shelter.

Terra Amata
This website gives more information about Terra Amata.

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Evidence from sites in other parts of the world reinforces the idea that *H. erectus* was a skilful hunter, employing a variety of techniques to capture game. At Olorgesailie, in south-western Kenya, the site of a massive slaughter of baboons has been located. This hunt must have been organised well in advance, as stones and tools had been carried to the site from up to 33 kilometres away. Organisation like this also indicates that, half a million years ago, *H. erectus* was capable of logical thought and had the ability to communicate and work with others in an organised and efficient manner.

In Spain, at Torralba and Ambrona, evidence indicates that hominins lit fires to drive elephants into swamps where they were trapped and butchered. It appears that this driving technique was also employed at Olduvai Gorge, in Africa, to trap antelopes and pigs. Once captured, the animals were butchered using tools made from bone and stone. The butchery marks on the surface of fossil bones indicate that, as time passed, *H. erectus* became more systematic in the use of tools. This suggests an increasing commitment to routine meat eating.

Tool-manufacturing sites were also found in France, and they included tools made from both stone and bone. However, no fossil hominins were found. The remains of animals indicated that hunting was important, and the predominance of deer bones suggested that the inhabitants preferred this type of meat. The presence of some fish bones indicated that these hominins also fished from time to time.

The life of *H. erectus* was significantly influenced by the use of fire. It was the first step towards manipulation of the environment to suit human needs. Fire helped keep predators away, gave warmth and light at night, and may have been used to stampede animals. The warmth from a fire would have been important for migrating groups moving into Europe and Asia during the bitter cold of the ice ages. Fire also enabled cooking, which increased the range of foods that could be eaten by improving flavour and digestibility. It would also have made some foods safer to eat, either by destroying the early stages of parasites such as tapeworms, which may be present in meat, or by detoxifying some plant foods.

FIGURE 13.22 The production of flake tools: **a** and **b** show preparation of the core, and **c** shows how a large number of flakes can be produced

Cultural changes such as the use of fire and the manufacture of tools would have influenced the social organisation of *H. erectus*. Greater emphasis must have been placed on mutual cooperation, and a complex society began to be established in which the care of the young would have gradually become increasingly important. A relatively complex spoken language could also have arisen by this time, but this is, of course, impossible to establish from the fossil record.

Tool use by *Homo neanderthalensis*

By the time of later hominins such as *Homo neanderthalensis*, further cultural advances had greatly diminished the importance of the environment in determining how and where they lived. Tool making now involved the production of stone flakes that could then be trimmed to form various cutting, scraping, piercing and gouging tools. Commonly referred to as the '**Mousterian industry**', after Le Moustier in France where the first flake tools were found, these tools showed a cultural advance over the Acheulian hand axe. A piece of stone was first trimmed into a disc-shaped core, and then struck by another piece of stone to produce the flakes that were flat on one side and had sharp edges. This technique is known as the **Levallois technique**. It is a slow, labour-intensive process that requires planning and foresight.

Levallois technique
This website includes an animation of the Levallois technique.

The flake tools could also be joined on to a handle, spear or arrow in a process called **hafting**. This broadened the use and increased the effectiveness of the tools. For example, spears could be used for hunting larger animals, hand axes could be used for cutting up animals or wood, while other tools could be used to make tools.

The Lavallois technique and hafting required planning and the ability to foresee possible outcomes. This indicates a significant development in the cognition of the species. This behavioural evolution coincides with the evolutionary trend in increased cranial capacity, which is likely possible due to the increased importance of meat in the diet.

Flake tools enabled people living in colder climates to become good clothes makers. Numerous scraping tools for preparing animal hides have been found at Neanderthal sites.

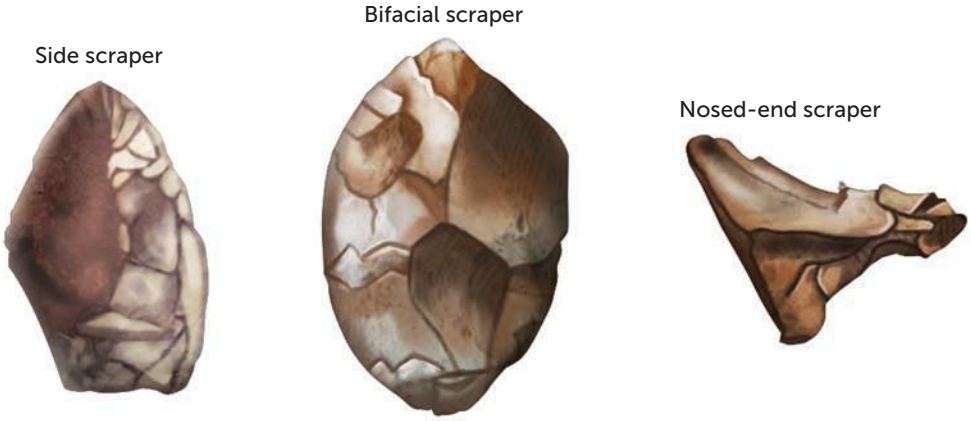

FIGURE 13.23
Neanderthal tools

The cultural advances of Neanderthals were not limited to tool making. There is strong evidence that Neanderthals buried their dead, leading to the suggestion that they believed in life after death. Ceremonial burial also seems to have been practised. At one site, the grave of a youth was surrounded by wild goat horns that had been thrust into the ground with the pointed ends downwards. At another site, a man had been buried on a bed of flowers. The shoulder blade, collarbone and upper right arm bone were all underdeveloped and there were no lower arm bones. Perhaps the man had been born with a withered right arm that had been successfully amputated above the elbow. It is likely that Neanderthals cared for disabled members of their group and had developed a social system for sharing food and other resources.

Tool use by *Homo sapiens*

Around 50 000 years ago, new technologies associated with modern humans – finer blades and projectile weapons – began to appear. Scientists can only speculate on what triggered this technological spurt. Some have suggested that there was a mutation that affected the brains of a group of anatomically modern humans living either in Africa or in the Middle East. This may have resulted in new neurological connections that gave them new abilities. Perhaps it permitted fully articulate speech, so these people could pass on information more efficiently.

Whatever the cause, around 40 000 years ago modern humans moved into Europe. They brought with them innovations such as clothing, which had been sewn, and better shelters. This allowed them to survive the cold of glacial Europe, previously the exclusive domain of Neanderthals. The populations of both peoples were small and scattered. But while modern humans began to thrive, Neanderthal populations gradually decreased.

These modern humans became well established in Europe and were the makers of blade tools – flakes of stone with roughly parallel sides. Known as the Cro-Magnon people, they had large brains housed in skulls that were long from front to back, similar to the present people of Western and Northern Europe. Cro-Magnon people were essentially hunters and gatherers, relying mainly on the hunting of herd animals that occupied the open plains. They mastered the art of hunting animals such as bison, mammoth and reindeer, often by stampeding them over cliffs or into narrow ravines. Besides being a source of meat, these animals also provided skins, which served as clothing or shelter.

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FIGURE 13.24
Aurignacian blade shown from three angles

There is evidence to suggest that the fat from these animals was used for oil lamps, and that their bones and ivory were used to make tools.

When the first fossil remains were found at Cro-Magnon in 1868, the tools found with them were similar to those found eight years earlier at Aurignac, which had become known as **Aurignacian tools**. These were blades made by removing long, flat rectangles from the core stone, which were easy to handle and effective in cutting.

Besides the Aurignacian tool culture, two other cultures are associated with later Cro-Magnon people: the Solutrean and the Magdalenian. The **Solutrean culture** was characterised by beautifully made willow-leaf and laurel-leaf points. These were made by carefully retouching blades produced from the original stone core by pressure flaking. The laurel-leaf point illustrated in Figure 13.25 must have taken many hours of intricate skill to produce and is thought to have been an ornament, or perhaps a symbol of the tool maker's craft, as it would have served little practical purpose.

The **Magdalenian** cultural period, which followed the Solutrean, was named after the rock shelter of La Madeleine in France. This culture is known for the dominance of bone and antler tools over those of flint and stone, and for the works of art that were produced during this period. The bone and antler tools were made using a burin, or chisel-like cutter, a tool used for the manufacture of other tools. This was a significant advance in tool making: humans had devised a tool for making other tools. To make the burin, a blade was shaped so that it had a sharp cutting point. With this, bone, antler and ivory could be cut to make a range of tools, from fine needles to barbed spear points and spear throwers.

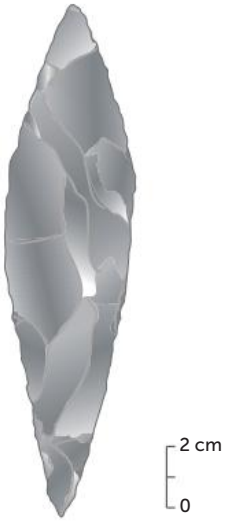

FIGURE 13.25
Solutrean 'laurel-leaf' blade

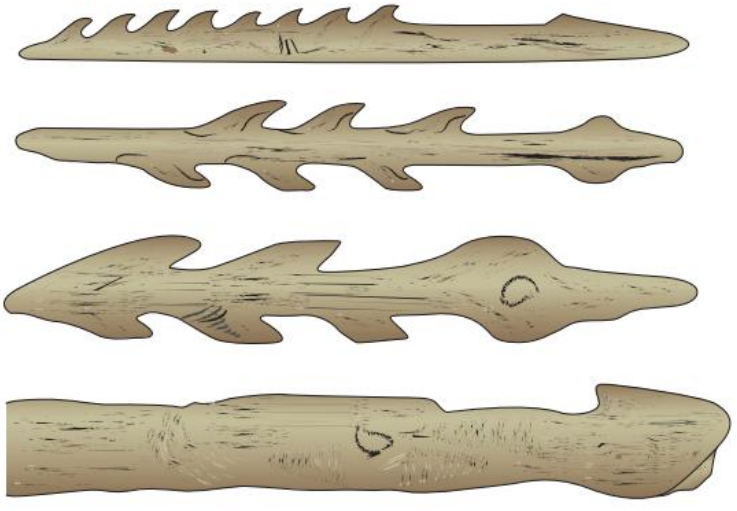

FIGURE 13.26
Magdalenian barbed points and spear thrower (bottom) made of bone or antler

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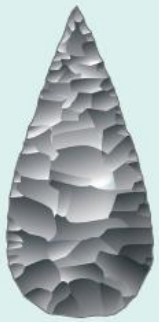
TABLE 13.5 Simplified table showing approximate age ranges and cultural periods of hominins

COMMON NAME	SCIENTIFIC NAME	APPROXIMATE AGE RANGE (YEARS BP)	TYPE, LOCATION	CULTURAL PERIOD	EXAMPLE
Australopithecines Handy man	*Australopithecus* sp. *Homo habilis*	2.6–1.7 million	Olduvai, Africa	Oldowan	
Homo erectus	*Homo erectus*	1.7 million–200 000	St Acheul, France	Acheulian	
Neanderthal	*Homo neanderthalensis*	200 000–40 000	Le Moustier, France	Mousterian – manufacture of flake tools	
Cro-Magnon	*Homo sapiens*	43 000–26 000	Aurignac, France	Aurignacian – manufacture of blade tools	
Cro-Magnon	*Homo sapiens*	22 000–19 000	Solutré, France	Solutrean – pressure flaking to retouch blades	
Cro-Magnon	*Homo sapiens*	18 000–12 000	La Madeleine, France	Magdalenian – predominance of bone and antler tools, and artwork	

Note: Tool cultures frequently persisted longer than shown; for example, a simple pebble tool would have been used by modern humans if this was all that was needed for a task. Age ranges are approximations, as different ages are associated with different sites.

Human evolution
This website provides a comprehensive description of human evolution, narrated by noted anthropologist Donald Johanson. Click on 'Launch the documentary'.

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Trends in tools

13.2 Cultural evolution of hominins

During the course of human evolution, some general trends are evident:

- increased manipulation of materials
- increased complexity of tools
- greater variety of materials being used to make tools
- improved workmanship and development of equipment needed to manufacture the tools
- increased specialisation of tools.

These trends, alongside structural changes during evolution, allow us to infer changes in lifestyles. Collaboration would have increased, requiring effective communication. Members within a group would have developed more specific roles and skills. Planning and creativity became important, and humans started manipulating the environment to meet their needs.

Activity 13.5
Examining chimpanzees, Neanderthals and humans

Key concept

During hominin evolution the use of tools also evolved from using tools that they found, such as pebbles, to sharpening edges to making flakes or structures from materials such as bone.

Questions 13.3

RECALL KNOWLEDGE

1. Describe Oldowan tools.
2. Name the species known to use Oldowan tools.
3. Which species was the first to use fire? List four different ways that fire could have been used.
4. What tool culture did Neanderthals use?
5. Name and describe the tools used by the Cro-Magnon.
6. Explain how the use of tools from the following cultures are related to the changes in cranial capacity of hominins: Oldowan, Acheulian, Mousterian, Aurignacian, Solutrean and Magdalenian.

APPLY KNOWLEDGE

7. State the name of the tool culture of the tool shown below. Justify your answer.


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8. Explain how Mousterian tools differ from Acheulian tools.
9. We can say that the environment influenced *Homo habilis*, but *Homo sapiens* influenced the environment. Relate this statement to the tools used by the two species.

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CHAPTER 13 ACTIVITIES

ACTIVITY 13.1 Investigating cranial capacity and phylogenetic trees

The subfamily Homininae includes humans, chimpanzees and gorillas, and their extinct ancestors. One of those extinct ancestors was *Ardipithecus ramidus*, who lived around 4.4 to 4.2 million years ago and who many scientists believe gave rise to the australopithecines and therefore could be a direct ancestor of modern humans. Even if *A. ramidus* is not on our direct evolutionary line, it must have been closely related to the direct ancestor, and was probably similar in appearance and adaptation.

Estimates of the cranial capacity of *A. ramidus* are between 300 and 350 cm^3, similar in size to modern female chimpanzees.

In this activity, you will use the information on cranial capacity in Table 13.1 to construct a phylogenetic tree of the hominins.

What to do

1. Assume that *A. ramidus* is the common ancestor of all the other species.
2. Consider which species may have become extinct and which species may have evolved into one or more other species. Draw up a phylogenetic tree to show the possible evolutionary relationships between the species in the table. Remember, there is no such thing as a correct tree. Scientists themselves cannot agree on all the relationships.
3. Once you have constructed your tree, go to the 'Understanding evolution' weblink to see how your tree compares with the information provided on that page.

Understanding evolution

SOUTHERN Biological Developed exclusively by Southern Biological

ACTIVITY 13.2 Investigating hominid skulls

Aim

To analyse various hominid/primate skulls

This is an excellent opportunity for you to explore various anatomical adaptations that have emerged in hominids over their evolution.

Time requirement: 45 minutes

You will need

Pan troglodytes (chimpanzee) (modern) skull; Gorilla (gorilla) (modern) skull; *Homo sapiens* (human) (modern) skull; *Homo neanderthalensis* (Neanderthal man) (120 000–30 000 years ago) skull; *Homo erectus* (upright man) (2.0 million years ago) skull; *Australopithecus boisei* (2.3–1.2 million years ago) skull; *Australopithecus afarensis* ('Lucy') (4.0 million years ago) skull; tape measure (in millimetres)

Risks

WHAT ARE THE RISKS IN THIS INVESTIGATION?	HOW CAN YOU MANAGE THESE RISKS TO STAY SAFE?
Skulls may have sharp edges.	Handle with care and do not run fingers over skull teeth.

What to do

Examining the braincase

1. Examine the frontal bone (forehead) of each of the skulls and determine if they appear more vertical or flatter. Ensure the skull eyes are oriented forward while doing this.
2. Examine above the orbital and determine if a supraorbital (brow ridge) is present. If so, see if the brow ridge is continuous or divided in the middle.
3. Measure the width of the braincase at the widest point. Make all measurements in millimetres.
4. Look for evidence of a sagittal crest running lengthwise along the midline of the top of the skull. Identify if it is prominent, present or absent.
5. Measure the distance between the front teeth and the front ridge of the foramen magnum.
6. Examine behind the ear of the skull and determine if the mastoid process is fairly flat or noticeably protruding.
7. Record the results of your observations by copying and completing Table 1.

Examining the facial structure

1. Position the skull so that it is facing you. Examine the nasal bones. Identify whether they are flat or protruding.
2. Measure the maximum breadth (width) of the nasal opening.
3. Measure the maximum height of the nasal opening.
4. Starting at the outside of the back molars, measure the width of the maxilla (the upper jaw).
5. The bizygomatic breadth is the width of the face from the widest part of one zygomatic arch to the widest part of the other zygomatic arch. Measure this distance.
6. Record the results of your observations by copying and completing Table 2.

Examining the dentition (teeth)

1. Examine the dental arcade (the shape made by the rows of teeth in the upper jaw). Observe the teeth towards the back and identify whether the teeth on each side of the jaw are parallel or diverging.
2. Reposition the skull so that you are viewing it from the side. Examine the incisors and identify if they are vertical or angled forward.
3. Measure the width of the incisors on the left side of the jaw and then measure the incisors on the right side of the jaw. Add the width of all incisors together to determine the combined width.
4. Examine the maxilla (upper jaw) and mandible (lower jaw) together. Identify whether the canine teeth project above or below the chewing surfaces of the other teeth.
5. See if you can identify a canine diastema (a gap on the medial side of the canine).
6. Measure from the back of the last molar to the front of the first premolar on the left side of the jaw. This will give you a measurement of the chewing surface of the teeth.
7. Record the results of your observations by copying and completing Table 3.

Studying your results

1. Copy and complete the tables below. Include a row for each specimen.

TABLE 1 Examining the braincase

SPECIMEN	FOREHEAD	BROW RIDGE (PRESENCE)	BROW RIDGE (CONTINUOUS OR DIVIDED)	BRAINCASE	SAGITTAL CREST	FORAMEN MAGNUM	MASTOID

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TABLE 2 Examining the facial structure

SPECIMEN	NASAL BONES	NASAL OPENING WIDTH	NASAL OPENING HEIGHT	MAXILLA WIDTH	BIZYGOMATIC BREADTH

TABLE 3 Examining the dentition (teeth)

SPECIMEN	DENTAL ARCADE	INCISORS	INCISORS WIDTH	CANINE	DIASTEMA	CHEWING SURFACE

2 Draw a graph of one characteristic (e.g. presence of brow ridge) from each table. Draw 'specimen' on the x-axis and arrange in order from great apes to modern humans.

Discussion

1 The canine teeth have drastically reduced in size from great apes to modern humans. Explain why this might be.
2 Explain why the face has become progressively flatter over the evolution of hominids.
3 Describe how the position of the foramen magnum relates to body posture and locomotion.
4 Certain areas of the braincase enlarged before others in our evolution. Describe how the areas enlarged throughout our evolution.
5 What traits differentiate modern apes and modern humans?
6 Using your measurements and the facial features you observed as evidence, do you think modern humans or modern apes are more closely related to extinct hominids? Explain your answer.
7 Imagine you found the remains of a skull that only contained the mandible. Is this enough evidence to determine if it belonged to a modern human, early hominid or ape? Explain your answer.

ACTIVITY 13.3 Investigating evidence for human evolution

Eugène Dubois was an anatomist who enlisted in the Dutch army so that he could go to Sumatra (in Indonesia) to look for fossils of human ancestors. Remarkably, Dubois found what he was looking for: a tooth, part of a skull and a thighbone that were clearly not from modern humans.

Dubois' discoveries generated great interest in human origins and raised awareness of the importance of fossil material. One fossil that proved to be significant was taken to the Australian anatomist Raymond Dart, who was working at the University of Witwatersrand in Johannesburg in the 1920s.

What to do

Use a variety of research techniques to investigate the evidence for human evolution that Dubois and Dart discovered, and to answer the following questions.

1 What fossils were discovered?
2 Where and when were the fossils found?
3 What was the scientific name given to the fossil finds at the time of their discovery?
4 What was the significance of the finds at the time? Did they raise any controversy in the scientific community?

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5 What is the significance of the fossils today, given that much more fossil evidence is available for study?
6 Other scientists who made significant contributions in the early days of the search for human origins were Robert Broom and Louis and Mary Leakey. Research the work of each of these people.

ACTIVITY 13.4 Are humans unique?

Modern humans like to think of themselves as unique. We consider ourselves to be different from (and perhaps superior to) all other species of animals. But are we unique? What separates us from other animals, especially the other primates?

With a partner, try to draw up a list of features that are unique to humans. Consider all aspects of humanity in your discussion – physical characteristics, behaviour, human achievements and others. Do some of the features selected follow an evolutionary trend? Are these features likely to evolve further in the future?

Have a class discussion of the lists proposed by the various pairs in the class and try to agree on a class list. Be prepared to criticise others but do so in a constructive way. It is more important to be involved in actively thinking about the topic than in arriving at a correct answer. In fact, there may be very few points on which the whole class will agree.

ACTIVITY 13.5 Examining chimpanzees, Neanderthals and humans

This activity will enable you to use knowledge gained to examine the relationship between chimpanzees, Neanderthals and modern humans. The following table indicates the number of nucleotide (or base) differences between a region of mitochondrial DNA in two chimpanzees, a Neanderthal and two humans.

	HUMAN 2	CHIMPANZEE 1	CHIMPANZEE 2	NEANDERTHAL
Human 1	15	77	76	20
Human 2		79	80	27
Chimpanzee 1			23	72
Chimpanzee 2				71

What to do

Answer the questions below. As you answer the questions, refer to the table and to previous chapters where necessary.

1 Based on the information in the table, which individual is most closely related to the Neanderthal and which is the least closely related?
2 The Neanderthal mitochondrial DNA was extracted from a fossil 25 000 years old. What other information obtained from the fossil would be valuable in determining the evolutionary relationships of the Neanderthal with chimpanzees and humans?
3 What dating methods could be used to determine the absolute age of the Neanderthal fossil?
4 What methods could have been used to determine a relative age for the Neanderthal fossil?
5 Use the data to draw a phylogenetic tree for these species.

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CHAPTER 13 SUMMARY

- Humans are hominins, being bipedal walkers and having less hair and a greater development of brain, speech and sexual characteristics than other hominids.
- During the evolution of hominins, the cranial capacity has gradually increased. There has also been an increase in the number of convolutions of the cerebral cortex and the size of the frontal lobe. This corresponds to a reduction in prognathism and the development of a forehead.
- Early hominins had a lower jaw and a face similar to the great apes. During evolution, the teeth became smaller. As they take up less space, this results in a flatter face.
- Fossil evidence of *Australopithecus afarensis* and *Australopithecus africanus* includes the Taung skull, Laetoli footprints and 'Lucy'.
- Australopithecines had short canines and a lack of diastema, with the teeth arranged in a parabolic shape. They had a low forehead and a projecting lower jaw. Their average cranial capacity was 480 cm^3. Their foramen magnum was more central than in other apes, and the skull more rounded at the back. Australopithecines were bipedal, with a non-opposable big toe and an 'S'-shaped spine. The fingers were heavily built and more suitable for a power grip than a precision grip.
- *Australopithecus afarensis* existed 3.9–2.8 million years ago, earlier than *Australopithecus africanus*, who existed 3.2–2.0 million years ago. They showed fewer evolutionary changes.
- *Paranthropus robustus* are thought to form a branch in hominin evolution, living 1.8–1.2 million years ago. They were robust with a large sagittal crest with strong chewing muscles and molars. They had a larger cranial capacity, with an average of 520 cm^3 and a wide, dish-shaped face with less prognathism.
- *Homo habilis* had a larger brain (610 cm^3) and smaller teeth than the australopithecines. When compared to the australopithecines, their skulls were rounder, the foramen magnum central, the dental arcade rounder and with less prognathism. The arms of *Homo habilis* were long and the legs short. The fingers were slightly curved, indicating a power grip. However, they were also capable of a precision grip.
- *Homo erectus* showed features more similar to modern humans than previous species, showing further evolutionary changes. Their cranial capacity was 1050 cm^3, their forehead low and sloping, and their jaw large, thick and rounded, without a chin. The molar teeth were smaller, indicating a diet similar to that of modern humans. *Homo erectus* were the first species to use fire.
- *Homo neanderthalensis* were an evolutionary branch who existed in Europe during the ice age. They were short in stature with a heavier build than modern humans. They had big faces, low but large skulls, heavy brow ridges and an occipital bun at the back of the skull. Their cranial capacity was larger than that of modern humans at 1485 cm^3. The face of Neanderthals showed greater prognathism than modern humans due to the nasal bones projecting forward.
- Cro-Magnon people were early *Homo sapiens*. Their skulls were shorter from front to back and higher than the skulls of Neanderthals. They also showed reduced brow ridges and prognathism, and brains averaging 1350 cm^3.
- Australopithecines used Oldawan tools – pebble tools that include choppers, scrapers, flakes and chisels. While they used the tools, they did not make or change them.
- *Homo habilis* continued to use Oldawan tools, but they sharpened or shaped them to be able to use them to hunt and scavenge.

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- *Homo erectus* used Acheulian tools – tools made by flaking the edges of stones in both directions – for hunting and fishing. *Homo erectus* also used fire and built shelters, allowing them to be independent of the environment.
- *Homo neanderthalensis* used Mousterian tools – flake tools produced by striking a disc of stone using the Lavallois technique. This allowed them to make clothes. They also attached the tools to handles, spears and arrows, increasing their use and effectiveness – for example, for hunting larger animals. Neanderthals buried and honoured their dead. It is also likely that they cared for less-abled individuals and shared food and resources.
- *Homo sapiens* developed fully articulate speech, leading to more effective communication. Their clothing and shelter became more sophisticated. The early *Homo sapiens*, the Cro-Magnon people, were hunters and gatherers. They used the animals for meat to eat, their fat for fuel, bones for tools, and the skin for clothing and shelter. Their tools were known as Aurignacian tools – tools made by removing long, flat rectangles from the core stone that are used as blades.
- Later Cro-Magnon people were part of the Solutrean culture. This is characterised by willow-leaf and laurel-leaf points. They were also part of the Magdalenian culture, which used bone and antlers that were modified by other tools. They also created art pieces.
- During evolution, tool use and construction has increased in complexity.

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CHAPTER 13 GLOSSARY

Acheulian tool A type of hand axe that was flaked all around the edges, first in one direction, and then in the other, until it formed a roughly two-faced lump, approximately teardrop in shape; associated with *Homo erectus*

Aurignacian tool The tool culture of stone, bone and antler associated with early Cro-Magnon people

Cranial capacity The volume of that part of the skull that is occupied by the brain

Cro-Magnon people The first anatomically modern people found in Europe

Cultural evolution Cultural development that occurs as a means of overcoming environmental and other challenges

Culture Anything that is learnt

Endocast An impression of the inside of the brain case, either artificial or natural, made of rock or some other solid material

Hafting The process of attaching a stone tool to a handle, spear or arrow

Home base A camp site to which prehistoric hunters brought back food for sharing with other members of their group

Lavallois technique The process of producing a flake from a stone core; flakes normally had a flat side and sharp cutting edges

Magdalenian A prehistoric culture known for a predominance of bone and antler over flint and stone tools, and for the works of art they produced

Mousterian industry Describes a tool characterised by the careful preparation of a stone core from which a large number of flakes could be removed; associated with *Homo neanderthalensis*

Oldowan tool A very simple tool made by removing several flakes from a stone; the stone tool culture of *Homo habilis*

Pebble tool A stone tool made by chipping flakes off a rounded pebble

Solutrean culture The stone tool culture characterised by pressure flaking stones to produce beautifully made willow-leaf and laurel-leaf points; associated with the later Cro-Magnon people

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CHAPTER 13 REVIEW QUESTIONS

Recall

1 Describe the main physical features of the genus *Australopithecus*.

2 Describe the features evident from a study of the skull of each of the following species.
- a *Australopithecus afarensis*
- b *Australopithecus africanus*
- c *Homo habilis*
- d *Homo erectus*
- e *Homo neanderthalensis*
- f *Homo sapiens*

3 List the differences between Neanderthals and modern humans.

4 Describe the physical appearance of Cro-Magnon people.

5 a What was the importance of meat eating to the future survival and evolution of the hominins?
- b How did tool manufacture and use contribute to this survival?

6 *Homo erectus* appears to be the first hominin to have used fire in a systematic way. List the ways in which fire could have improved their way of life, giving examples where appropriate.

7 Describe the significant cultural advance that occurred with the development of the Mousterian tool-making industry.

Explain

8 Describe the significance of the Laetoli footprints and explain why they were such an important discovery.

9 Explain how hafting changed the use of stone tools.

10 Most of the major changes in human evolution from *Homo erectus* to modern *Homo sapiens*, identifiable from fossil evidence, are confined to the head. Identify five of these changes and explain their significance.

Apply

11 What assumptions are made when scientists infer the degree of intelligence from the cranial capacity of a skull?

12 In the past, anthropologists have put a great deal of emphasis on the importance of the cranial capacity when defining the tribe Hominini. Does this seem reasonable, considering the hominins discussed in this and the previous chapter? What other physical features are important in a discussion of human evolution?

13 There is growing evidence that, like many of the other mammals, the pathway to modern humans may have many more species existing at a particular time than was once thought. If this is the case, how would it have been possible for closely related species to have lived on Earth at the same time? Describe a possible situation where three species of early *Homo* lived in the same region of Africa.

14 Describe the conditions that may have led to Neanderthals developing their characteristic anatomical features.

15 Compile a phylogenetic tree for the evolution of hominins from the early australopithecines to modern humans. List evidence in support of your evolutionary pathway and discuss any points of disagreement that others may have with it.

16 Why do scientists believe that the laurel-leaf blade may have been an ornament rather than a spear-point?

17 Australopithecines may have been the first hominins to manufacture tools for a specific purpose. Describe the significance of this development in food gathering for later hominin evolution.

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18 There is some speculation among scientists that the large brain of *Homo erectus* would have required offspring to be born at a very early stage to allow the passage of the large head through a relatively narrow birth canal. Discuss the implications that the care of helpless young would have had for the social behaviour of *Homo erectus*.

19 Briefly outline the technological advances in tool making from the early Oldowan industry to that of Magdalenian times.

Extend

20 Who was 'Lucy', and why is she such an important 'person' in present theories of hominin evolution?

21 For the past 100 000 years at least, hominins have adapted culturally to environmental change. Does natural selection affect cultural characteristics?

22 Evidence suggests that *H. erectus* used fire to illuminate caves and other forms of shelter. To use fire effectively, they must have developed ways of lighting a fire and maintaining it for long periods. Use references to find out how early hominins may have lit fires and kept them burning.

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ISBN 978-0170449168
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