BIOZONE

Student Workbook

YEAR 12 BIOLOGY



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Student Workbook

Previous annual editions 1996-2014
Twentieth edition 2015

ISBN 978-1-927173-97-8
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Published by **BIOZONE International Ltd**

Printed by REPLIKA PRESS PVT LTD using paper produced from renewable and waste materials

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www.**BIOZONE**.com.au



Cover Photograph

The smooth knob-tailed gecko (*Nephrurus levis*) is an Australian terrestrial desert species. It is named for its distinctive broad tail, which ends in a small knob. Smooth knob-tailed geckos can live for 10 years, grow to about 10 cm long, and weigh around 20 grams. They are relatively common and are found throughout the Australian deserts. The smooth knob-tailed gecko is nocturnal and rests in sandy burrows during the day. There are three subspecies.

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Note to the Student

This year's edition of Year 12 Biology has been written specifically to help you develop the skills and knowledge to achieve success in your biology programme. We have provided a wide range of activities that will enable you to build on what you know already, explore new topics, work with your classmates, and practise your skills in data handling and interpretation and in answering free response questions as required in examinations. We hope that you find the workbook valuable and that you make full use of its features.

To use this workbook most effectively, take note of the features outlined in this introduction. Understanding the activity coding system and using the external resources we provide will help to scaffold your learning so that you understand the principles and processes involved in each topic of study.

Look out for these features and know how to use them:

- A guide to answering essay style questions is provided at the end of this introduction. Use it to help you when completing the synoptic questions that conclude each area of study.
- The chapter introduction provides you with the learning aims for the topic. Use the check boxes to identify and then mark off the objectives as you complete them. The chapter introduction also provides you with a list of key terms for the chapter, from which you can construct your own glossary as you work through the activities. See *Using this Workbook* on page 1 for more information.
- The activities form the bulk of this workbook. They have codes assigned to them according to the skills they emphasise. Each has a short introduction to the topic and provides some relevant background. Most of the information is associated with pictures and diagrams, and your understanding of the content is reviewed through the questions. Some of the activities involved modelling and group work.
- The free response questions allow you to use the information on the page to answer a question about the core ideas of the activity. They may require you to apply your understanding to a new situation where the same principles operate.
- A coding system on the page tab identifies the type of activity. For example, is it primarily information-based (*KNOW*), or does it involve modelling (*PRAC*) or data handling (*DATA*)? A full list of codes is given on page 1 but the codes themselves are relatively self explanatory.
- ***Link*** tabs at the bottom the activity page identify connections across the curriculum and pages covering similar or related concepts in different situations.
- The **Weblinks** resource provides you with external, online support material in the form of animations, video clips, and quizzes. Bookmark the page (see details on page 2) and visit it frequently as you progress through the workbook.
- ***VOCAB*** activities enable you to test your understanding of the key terms used in the chapter. Being able to define scientific terms accurately and use them appropriately in your exam answers will help you to achieve better results in exams.

Meet the writing team

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Founder & CEO

Thanks also to:

The staff at BIOZONE, including Gemma Conn and Julie Fairless for design and graphics support, Paolo Curray for IT support, Debbie Antoniadis and Tim Lind for office handling and logistics, and the BIOZONE sales team.

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Using This Workbook

This workbook is designed to increase your understanding and enjoyment of biology. It addresses the requirements of the Year 12 syllabuses for biology in Australia, including Victoria (VCE), Western Australia, Queensland, and South Australia. Use it as a stand-alone or complementary resource for your course work and home study.

Introduction

- A check list of learning aims
- A list of key terms

Activities

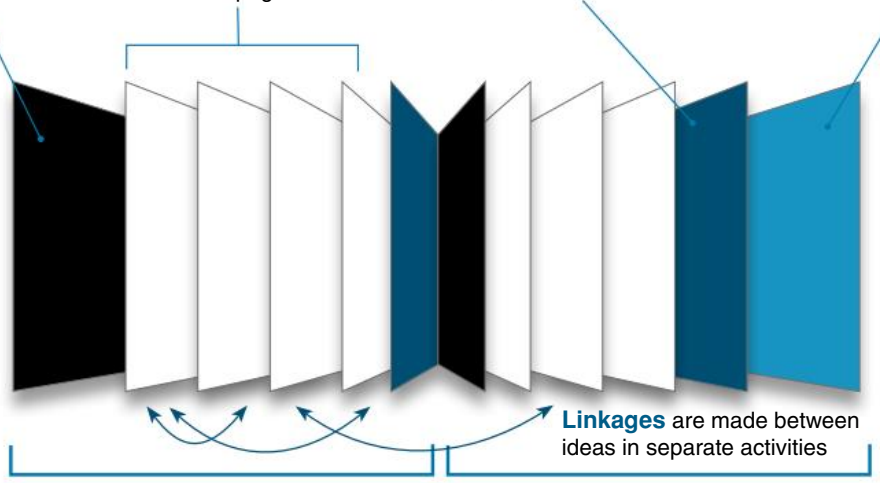
- The KEY IDEA provides your focus for the activity
- Annotated diagrams help you understand the content
- Questions review the content of the page

Literacy

- Most chapters have a literacy activity based on the introductory key terms list
- Several types of activities test your understanding of biological terms

Synoptic Question

- Synoptic questions conclude the four broad areas of study covered in the workbook.
- These enable you to practise your written exam skills



Structure and organisation of chapters

- The activities make up most of this workbook. Each one has a similar structure and they are organised through the chapter in a way that unpacks the material in a series of steps.

Activity number

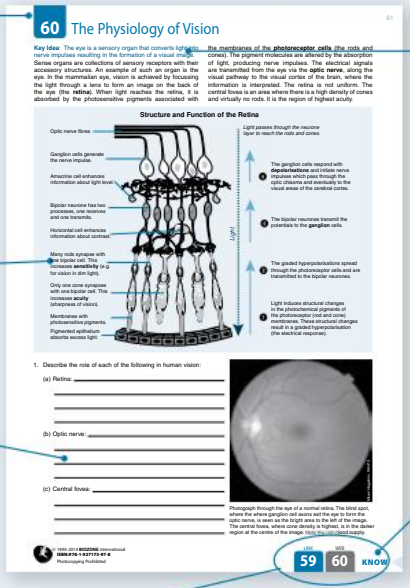
Activities are numbered to make it easy to navigate through the workbook and to find related activities.

Content explained

The main ideas of the activity are explained with short paragraphs and clear, informative diagrams, illustrations, and photos.

Direct questioning style

The questions help you consolidate your understanding of biological vocabulary, processes, and principles around the key idea.



Key Idea

The key idea provides the message for the activity. It helps you to understand what is important on the page.

Page tabs

The **page tabs** at the bottom of an activity page indicate the type of activity, related activities and external weblinks.



Link

Connections are made between activities in different sections of the syllabus that are related through content or because they build on prior knowledge.

Weblinks

Bookmark the weblinks page:
Bookmark the weblinks page:
www.biozone.com.au/weblink/au12-3978/

Access the external URL for the activity by clicking on its number

Activities are coded

COMP = comprehension of text
DATA = data handling and interpretation
KNOW = content you need to know
PRAC = a paper practical or a practical focus
REFER = reference - use this for information
TEST = test your understanding
VOCAB = learning your biological vocabulary

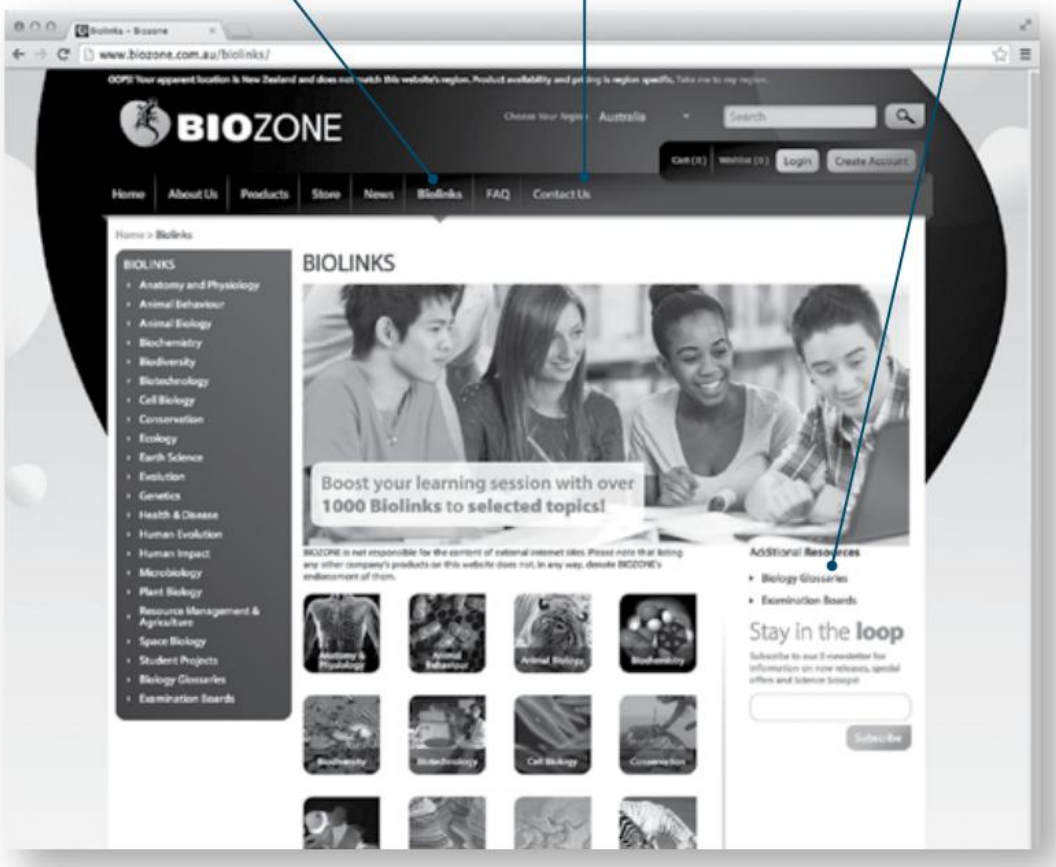
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Using BIOZONE's Website

Access the **BIOLINKS** database of web sites directly from the homepage of our new website. Biolinks is organised into easy-to-use sub-sections relating to general areas of interest. It's a great way to quickly find out more on topics of interest.

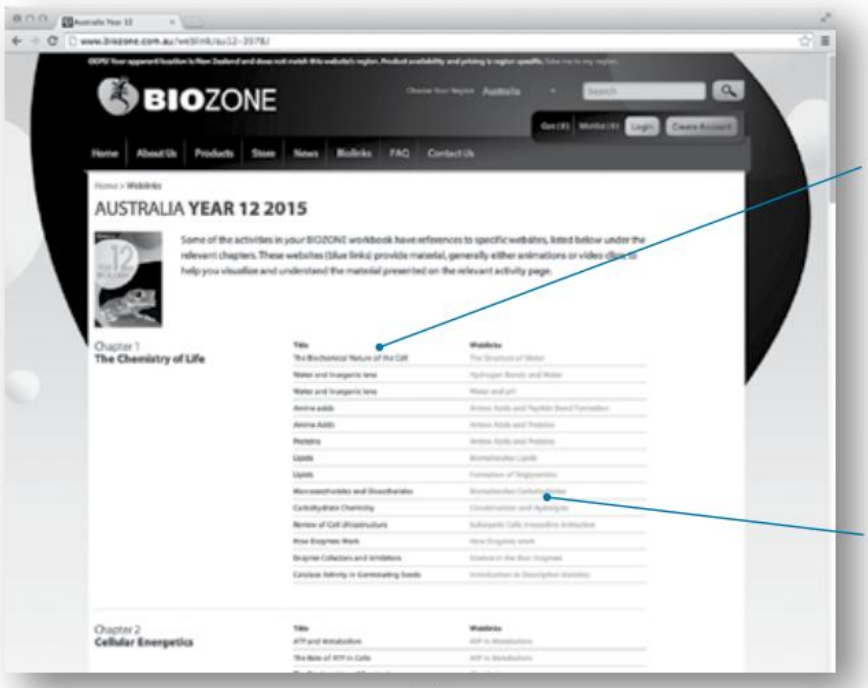
Contact us with questions, feedback, ideas, and critical commentary. We welcome your input.

Make use of additional resources, such as course information through your own state's examination board.



Weblinks: www.biozone.com.au/weblink/au12-3978/

***Bookmark the weblinks page by typing in the address**. It is not accessible directly from BIOZONE's website*



Throughout this workbook, there are references to **Weblinks** that are relevant to the activity on which they are cited. These are video clips, animations, or quizzes that support your understanding of the concepts on the page.

Weblink: Provides a link to an **external web site** with supporting information for the activity. If there are any additional activities, these are also listed here as links.

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Answering Exam Questions

- The external exams require you to demonstrate your understanding of a particular concept by providing a written paragraph or essay.
- Open answer questions (meaning there is no definitive answer) are designed so that you can demonstrate your level of understanding. The question may give you some guidance as to what you should include in your answer, such as definitions of certain terms or advice to provide specific examples.
- In order to gain the highest possible mark in these questions, you need to lay out your answer in a clear and logical way so that the examiner can easily see how you have demonstrated your understanding of the topic.
- The difference between you obtaining an a low, mid, or high grade depends on how well you demonstrate your understanding of a concept.
  - Defining, drawing, annotating, or giving a description demonstrates a basic understanding of the material.
  - Explaining how a process works, why it works, and how changes to it may affect an outcome shows a deeper understanding of how the system works in that situation.
  - Linking biological ideas, comparing and contrasting, analysing, or justifying shows both a deep understanding and an ability to translate that understanding to a new situation.
- The following example question shows how an answer can be built up from a simple definition, through explanation, to comparisons and linking of ideas.

Gene mutations may affect the protein formed during gene expression. Using an example, discuss the significance of mutations and the effect they may have on the fitness of an organism:

A gene mutation is a change to the base pairs in a DNA sequence that makes up a gene.

The term gene mutation is defined

Mutations can involve changes to a single nucleotide (called point mutations), or changes to a triplet by the insertion or deletion of a nucleotide. These are called frame shift mutations.

Different types of gene mutations are described.

Mutations can affect the protein formed, but some types of gene mutations are more likely to significantly alter proteins than others. Frame shift mutations can cause significant changes because the instructions for making a protein can be changed, producing a completely different amino acid sequence or a protein that is partly completed. Point mutations may also result in a different amino acid being inserted, but sometimes the mutation still codes for the same amino acid and the protein produced is the same.

The significance of different types of gene mutations is discussed.

Mutations can be harmful, beneficial, or have no apparent effect. The type of mutation can affect the fitness of an organism in its current environment. Fitness is a measure of an organism's ability to survive and reproduce (contribute genes to the next generation).

Effect of mutations on fitness is explained.

The term fitness is defined

An example of a beneficial mutation improving fitness is antibiotic resistance in bacteria. Some gene mutations in bacteria confer resistance to specific antibiotics. Bacteria with the beneficial gene mutation are not killed by the antibiotic and survive to reproduce. When they reproduce, the gene is passed on to the next generation and resistance to that particular antibiotic spreads quickly through the population. In contrast, bacteria without the gene mutation have decreased fitness for that particular antibiotic and are killed by it (so can not reproduce).

Example used to link the biological idea of how a beneficial mutation affects fitness

Comparing and contrasting the effect of a mutation on a population's fitness

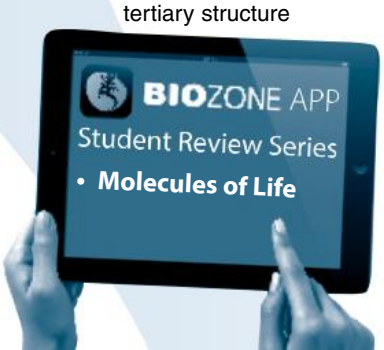
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QLD As required
VCE Unit 3 (1)
WA ATAR Unit 2
SA Theme M, C

The Chemistry of Life

Key terms

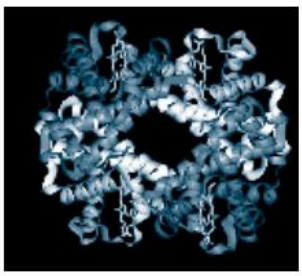
activation energy
active site
amino acid
anabolic reactions
carbohydrate
catabolic reactions
catalyst
cholesterol
coenzyme
cofactor
condensation
denaturation
disaccharide
endergonic reaction
enzyme
enzyme inhibition
exergonic reaction
fibrous protein
hydrolysis
globular protein
hydrogen bond
induced fit model
inorganic ion
lipid
lock-and-key model
macromolecule
monomer
monosaccharide
nucleic acid
optimum (for enzyme)
polymer
polysaccharide
primary structure
protein
proteome
quaternary structure
secondary structure
tertiary structure

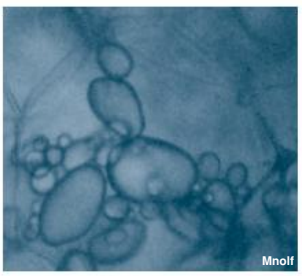


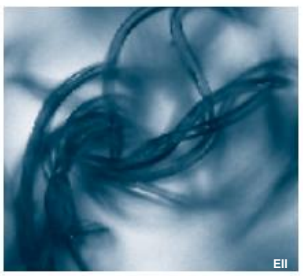
The chemical nature of cells

Learning aims and skills

		Activity number
☐	1 Describe the importance of inorganic ions and organic molecules (e.g. carbohydrates, proteins, lipids, and nucleic acids) in biological systems.	1 3
☐	2 Describe the structure of water. Explain the physical and chemical properties of water that are important in biological systems.	2
☐	3 Describe examples of monosaccharides and the biological significance of monosaccharide isomers. Describe how glycosidic bonds are formed and broken in the synthesis (by condensation) and hydrolysis of a disaccharide and a polysaccharide (e.g. amylose).	4 5
☐	4 Compare and contrast the structure of glucose polymers: starch, cellulose, and glycogen. Relate the structure to their biological function in each case.	6
☐	5 Describe the general structure of an amino acid. Explain how dipeptides and polypeptides are formed and broken apart by condensation and hydrolysis.	7
☐	6 Explain what is meant by the proteome. Describe the structural and functional diversity of proteins. Distinguish between the primary (1°), secondary (2°), and tertiary (3°) structure of a protein and relate these to function. Explain quaternary (4°) structure with reference to haemoglobin.	8
☐	7 Describe how proteins are modified after production. Describe the role of cellular organelles in the packaging and transport of macromolecules.	9 10
☐	8 Describe examples of nucleic acids and their roles in biological systems. Describe the role of condensation in forming nucleic acids (e.g. DNA).	11
☐	9 Describe the structure of triglycerides (fats and oils), phospholipids, and cholesterol. Relate the structure to their biological function in each case.	12
☐	10 Describe simple tests for sugars, starch, proteins, and lipids.	13







Review of cell ultrastructure

Learning aims and skills

		Activity number
☐	11 Recall the ultrastructure of plant and animal cells. Explain how different cell fractions may be separated by ultracentrifugation.	14-16
☐	12 Recall the structure of cell membranes and their role in cell structure and function.	17

Enzymes and metabolism

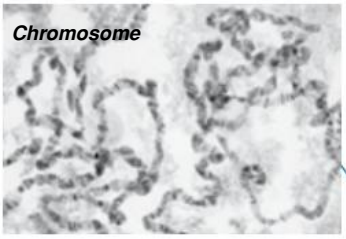
Learning aims and skills

		Activity number
☐	13 Describe the properties and mode of action of enzymes, including the role of the active site, specificity, and activation energy. Explain models for enzyme function and outline role of enzymes in anabolic and catabolic reactions.	18 19
☐	14 Describe the effect of pH, temperature, substrate concentration, and enzyme concentration on enzyme activity. Recognise that enzymes can be denatured.	20 23
☐	15 Using examples, explain the role of cofactors and inhibitors on enzyme activity.	21 22

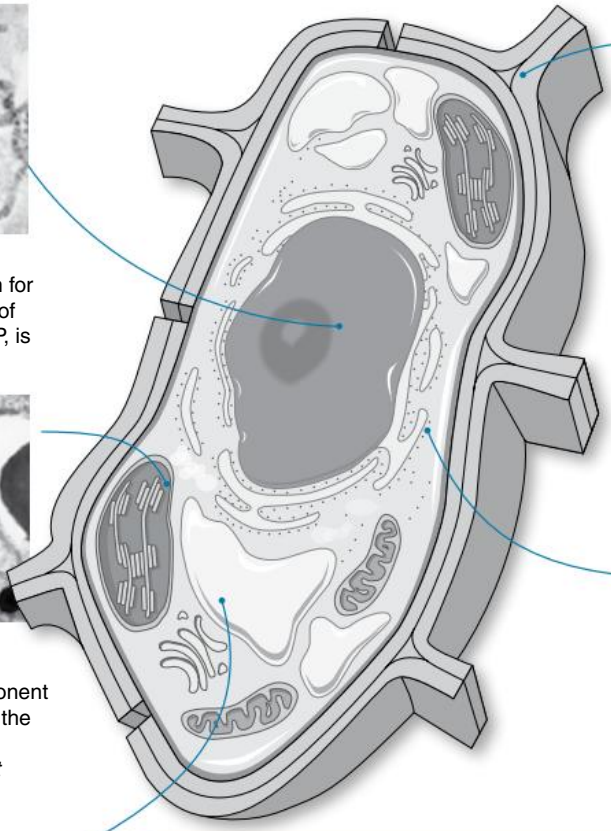
1 The Biochemical Nature of the Cell

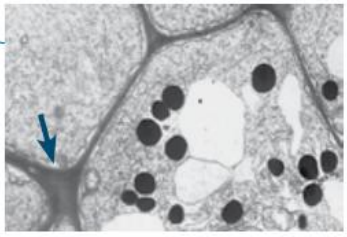
Key Idea: The main components of a cell are water and compounds of carbon, hydrogen, nitrogen, and oxygen.

Water is the main component of organisms, and provides an equable environment in which metabolic reactions can occur. Apart from water, most other substances in cells are compounds of carbon, hydrogen, oxygen, and nitrogen. The combination of carbon with other elements provides a huge variety of molecular structures, called **organic molecules**. The organic molecules that make up living things can be grouped into four broad classes: carbohydrates, lipids, proteins, and nucleic acids. In addition, a small number of **inorganic ions** are also components of larger molecules.

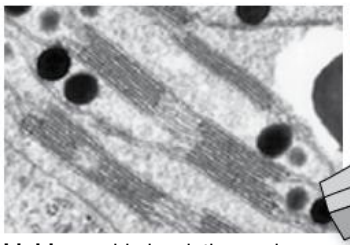


Nucleotides and nucleic acids Nucleic acids encode information for the construction and functioning of an organism. The nucleotide, ATP, is the energy currency of the cell.

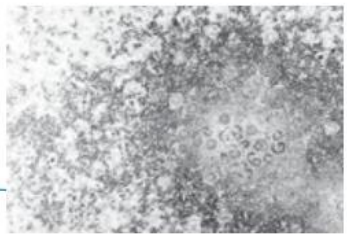




Carbohydrates form the structural components of cells, e.g. cellulose cell walls (arrowed), they are a store of energy, and they are involved in cellular recognition.



Lipids provide insulation and a concentrated source of energy. Phospholipids are a major component of cellular membranes, including the membranes of organelles.
Above: Thylakoid of a chloroplast



Proteins may be catalytic (enzymes), structural (e.g. collagen in skin, proteins in ribosomes), or they may be involved in movement, message signalling, internal defence and transport, or storage.
Above: ribosomes in translation

Water is a major component of cells: many substances dissolve in it, metabolic reactions occur in it, and it provides support and turgor.

Inorganic ions are important for the structure and metabolism of all living organisms. An ion is simply an atom (or group of atoms) that has gained or lost one or more electrons. Many of these ions are soluble in water. Some of the inorganic ions required by organisms and their biological roles are listed in the table on the right.

Ion	Name	Example of biological roles
Ca^{2+}	Calcium	Component of bones and teeth, required for muscle contraction
Mg^{2+}	Magnesium	Component of chlorophyll, role in energy metabolism
Fe^{2+}	Iron (II)	Component of haemoglobin and cytochromes
NO_3^-	Nitrate	Component of amino acids
PO_4^{3-}	Phosphate	Component of phospholipids and nucleotides, including ATP
Na^+	Sodium	Component of extracellular fluid and needed for nerve function
K^+	Potassium	Important intracellular ion, needed for heart and nerve function
Cl^-	Chloride	Component of extracellular fluid in multicellular organisms

1. Describe the biologically important roles of each of the following molecules:

   (a) Lipids: _____

   (b) Carbohydrates: _____

   (c) Proteins: _____

   (d) Nucleic acids: _____

2. Using examples, distinguish between inorganic and organic molecules and their general roles in cells:

   _____

   _____

   _____

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2 The Role of Water

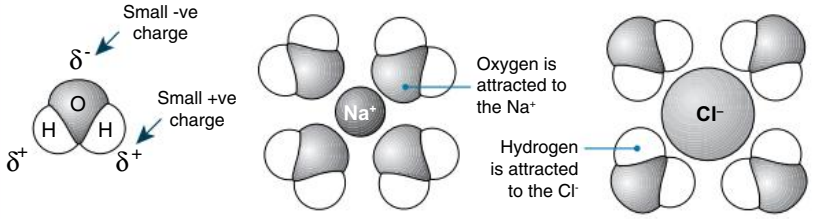
Key Idea: Water forms bonds between other water molecules and also with ions allowing it to act as a medium for the biological reactions of life.

Water typically makes up about two-thirds of any organism. Water is a liquid at room temperature and many substances dissolve in it. It is a medium inside cells and for aquatic life. Water takes part in, and is a common product of, many reactions. Water molecules are **polar** and have a weak attraction for each other and inorganic ions, forming large numbers of weak hydrogen bonds. It is this feature that gives water many of its unique properties, including its low viscosity and its chemical behaviour as a **universal solvent**.

Important Properties of Water



A lot of energy is needed to make water change state, so aquatic environments are thermally stable and sweating and transpiration cause rapid cooling.



Water molecule
Formula: H_2O

Water surrounding a positive ion (Na^+)

Water surrounding a negative ion (Cl^-)

The most important feature of water is its **dipole nature**. It has a small positive charge on each of the two hydrogens and a small negative charge on the oxygen.



Water is colourless, with a high transmission of visible light, so light penetrates tissue and aquatic environments.



Ice is less dense than water so ice floats, insulating the underlying water and providing valuable habitat.



Water has low viscosity, strong cohesive properties, and high surface tension. It flows freely through small spaces.

1. On the diagram above, showing a positive and a negative ion surrounded by water molecules, indicate the polarity of the water molecules (as shown in the example provided).

2. Explain the importance of the **dipole nature** of water molecules to the chemistry of life: _____

_____

_____

3. For (a)-(f), identify the important property of water, and describe an example of that property's biological significance:

   (a) Property important in the clarity of seawater: _____

   Biological significance: _____

   (b) Property important in the transport of water in xylem: _____

   Biological significance: _____

   (c) Property important in the relatively stable temperature of water bodies: _____

   Biological significance: _____

   (d) Property important in the transport of glucose around the body: _____

   Biological significance: _____

   (e) Property important in the cooling effect of evaporation: _____

   Biological significance: _____

   (f) Property important in ice floating: _____

   Biological significance: _____

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3 Organic Molecules

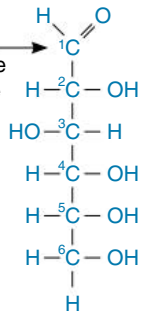
Key Idea: Organic (carbon-based) molecules make up most of the chemicals found in living organisms.

All life is based around carbon, which is able to combine with many other elements to form a large number of carbon-based (or organic) molecules. Specific groups of atoms, called functional groups, attach to a C-H core and determine the specific chemical properties of the molecule.

Some organic molecules are small and simple, containing only one or a few functional groups, while others are large complex assemblies called **macromolecules**. The organic macromolecules that make up living things can be grouped into four classes: carbohydrates, lipids, proteins, and nucleic acids. The diagram below illustrates some of the common ways in which organic molecules are portrayed.

Portraying Organic Molecules

The numbers next to the carbon atoms are used for identification when the molecule changes shape

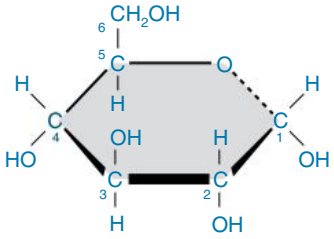


$C_6H_{12}O_6$

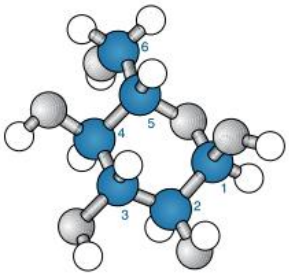
Glucose

Molecular formula

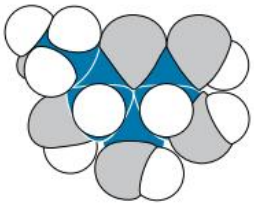
Structural formula
Glucose (straight form)



Structural formula
α-glucose (ring form)



Ball and stick model
Glucose



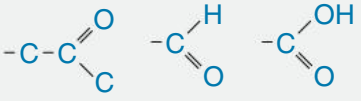
Space filling model
β-D-glucose

The molecular formula expresses the number of atoms in a molecule, but does not convey its structure. This is indicated by the structural formula.

A ball and stick model shows the arrangement of bonds while a space filling model gives a more realistic appearance of a molecule.

Examples of Organic Molecules

Organic (or biological) molecules may also include atoms other than carbon, oxygen, and hydrogen atoms. Nitrogen and sulfur are components of molecules such as amino acids and nucleotides. Some molecules contain the C=O (carbonyl) group. If this group is joined to at least one hydrogen atom it forms an aldehyde. If it is located between two carbon atoms, it forms a ketone.



Ketone **Aldehyde** **Carboxyl**



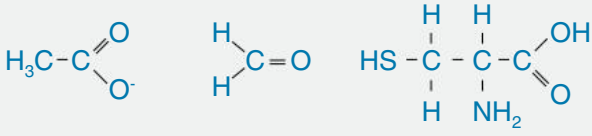
Acetate



Methanal



Cysteine



Key to Symbols

- Carbon
- Hydrogen
- Oxygen
- Nitrogen
- Sulfur

1. Which three main elements make up the structure of organic molecules? _____
2. Name two other elements that are also frequently part of organic molecules: _____
3. State how many covalent bonds a carbon atom can form with neighbouring atoms: _____
4. Distinguish between molecular and structural formulae for a given molecule: _____

_____

_____

5. What is a functional group? _____

_____

6. Classify methanal according to the position of the C=O group: _____
7. Identify a functional group always present in amino acids: _____
8. Identify the significance of cysteine in its formation of disulfide bonds: _____

_____

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4 Monosaccharides and Disaccharides

Key Idea: Sugars (monosaccharides and disaccharides) are simple carbohydrates with a role in providing energy and forming macromolecules, such as starch and glycogen.
Monosaccharide polymers form the major component of most plants (as cellulose). Monosaccharides are important as a primary energy source for cellular metabolism. Disaccharides are important in human nutrition and are found in milk (lactose) table sugar (sucrose) and malt (maltose).

Monosaccharides

Monosaccharides are used as a primary energy source for fuelling cell metabolism. They are single-sugar molecules and include glucose (grape sugar and blood sugar) and fructose (honey and fruit juices). The commonly occurring monosaccharides contain between three and seven carbon atoms in their carbon chains. Of these, the 6C hexose sugars occur most often. All monosaccharides are classified as reducing sugars (i.e. they can participate in reduction reactions).

Single sugars (monosaccharides)

Triose

C
C
C

e.g. glyceraldehyde

Pentose

e.g. ribose, deoxyribose

Hexose

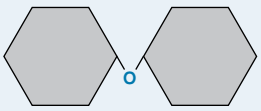
e.g. glucose, fructose, galactose

Disaccharides

Disaccharides are double-sugar molecules and are used as energy sources and as building blocks for larger molecules. The type of disaccharide formed depends on the monomers involved and whether they are in their α- or β- form. Only a few disaccharides (e.g. lactose) are classified as reducing sugars.

Sucrose = α-glucose + β-fructose (simple sugar in plant sap)
Maltose = α-glucose + α-glucose (a product of starch hydrolysis)
Lactose = β-glucose + β-galactose (milk sugar)
Cellobiose = β-glucose + β-glucose (from cellulose hydrolysis)

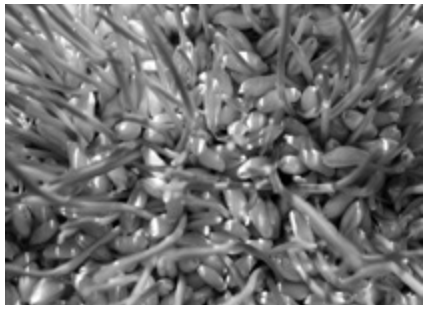
Double sugars (disaccharides)



Examples sucrose, lactose, maltose, cellobiose



Lactose, a milk sugar, is made up of β-glucose + β-galactose. Milk contains 2-8% lactose by weight. It is the primary carbohydrate source for suckling mammalian infants.



Maltose is composed of two α-glucose molecules. These germinating wheat seeds contain maltose because the plant breaks down their starch stores to use it for food.



Sucrose (table sugar) is a simple sugar derived from plants such as sugar cane (above), sugar beet, or maple sap. It is composed of an α-glucose molecule and a β-fructose molecule.

1. Describe the two major functions of monosaccharides:

   (a) _____

   (b) _____

2. The breakdown of a disaccharide into its constituent monosaccharide units is an enzyme catalysed hydrolysis (see opposite). For each of the following common dissaccharides, identify the enzyme responsible for the catalysis and the products of the hydrolysis, and describe an example of where this enzyme might naturally occur:

   (a) Lactose: Enzyme: _____ Products of hydrolysis: _____

   Found: _____

   (b) Maltose: Enzyme: _____ Products of hydrolysis: _____

   Found: _____

   (c) Sucrose: Enzyme: _____ Products of hydrolysis: _____

   Found: _____

3. Use your understanding of disaccharide chemistry to suggest how the digestive disorder lactose intolerance arises:

   _____

   _____

   _____

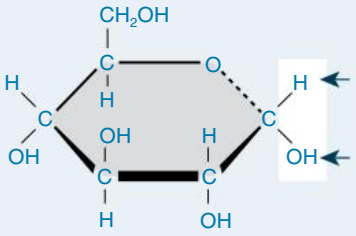
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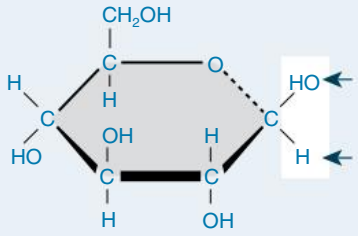
5 Carbohydrate Chemistry

Key Idea: Sugar monomers are linked by condensation reactions and broken into monomers by hydrolysis reactions. Sugar monomers are linked together by **condensation**, which results in the production of a water molecule. The reverse reaction, in which compound sugars are broken down into monosaccharides, is called **hydrolysis**. It splits polymers into smaller units by breaking the bond between two monomers. Hydrolysis literally means breaking with water, and so requires the addition of a water molecule to occur. Carbohydrates also exist as **isomers**. Isomers are compounds with the same molecular formula, but they have a different structural formula. Because of this they have different properties. For example, when α–glucose polymers are linked together they form starch, but β–glucose polymers form cellulose. In all carbohydrates, the structure is closely related to their functional properties.

Isomerism

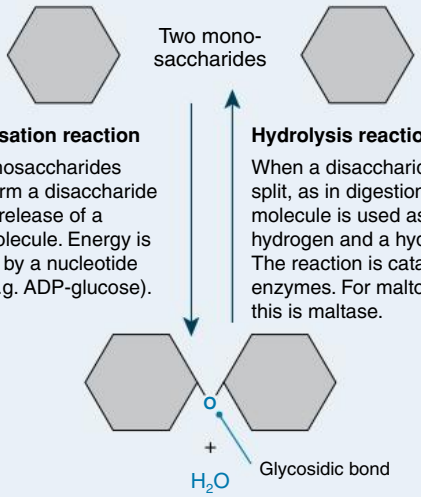
Compounds with the same chemical formula (same types and numbers of atoms) may differ in the arrangement of their atoms. Such variations in the arrangement of atoms in molecules are called isomers. In structural isomers (such as fructose and glucose, and the α and β glucose, right), the atoms are linked in different sequences. Optical isomers are identical in every way but are mirror images of each other.





Condensation and Hydrolysis Reactions

Monosaccharides can combine to form compound sugars in what is called a condensation reaction. Compound sugars can be broken down by hydrolysis to simple monosaccharides.



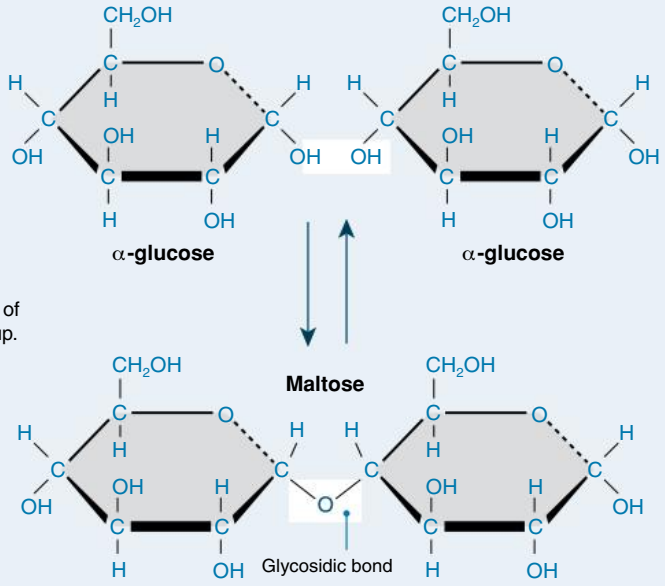
Condensation reaction

Two monosaccharides join to form a disaccharide with the release of a water molecule. Energy is supplied by a nucleotide sugar (e.g. ADP-glucose).

Hydrolysis reaction

When a disaccharide is split, as in digestion, a water molecule is used as a source of hydrogen and a hydroxyl group. The reaction is catalysed by enzymes. For maltose (right), this is maltase.

Disaccharide + water



Disaccharide + water

1. Distinguish between structural and optical isomers in carbohydrates, describing examples of each:

2. Explain how compound sugars are formed and broken down:

3. Using examples, explain how the isomeric structure of a carbohydrate may affect its chemical behaviour:

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6 Polysaccharides

Key Idea: Polysaccharides are straight or branched chains of many monosaccharides of the same or different types.

The most common polysaccharides (often called complex carbohydrates) are cellulose, starch, and glycogen. They contain only glucose, but their properties are very different. These differences are a function of the glucose isomer involved and the types of linkages joining them. Different polysaccharides, based on the same sugar monomer, can thus be highly soluble and a source of readily available energy or a strong structural material that resists being digested.

Cellulose

Cellulose is a structural material in plants and is made up of unbranched chains of β-glucose molecules held together by 1,4 glycosidic links. As many as 10,000 glucose molecules may be linked together to form a straight chain. Parallel chains become cross-linked with hydrogen bonds and form bundles of 60-70 molecules called **microfibrils**. Cellulose microfibrils are very strong and are a major component of the structural components of plants, such as the cell wall (photo, right).

Starch

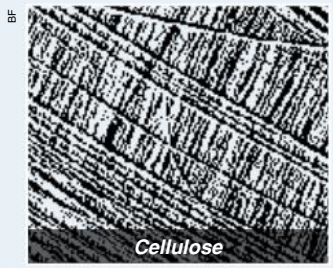
Starch is also a polymer of glucose, but it is made up of long chains of α-glucose molecules linked together. It contains a mixture of 25-30% amylose (unbranched chains linked by α-1,4 glycosidic bonds) and 70-75% amylopectin (branched chains with α-1, 6 glycosidic bonds every 24-30 glucose units). Starch is an energy storage molecule in plants and is found concentrated in insoluble starch granules within plant cells (see photo, right). Starch can be easily hydrolysed by enzymes to soluble sugars when required.

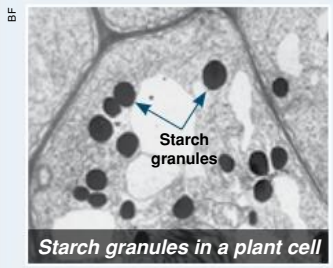
Glycogen

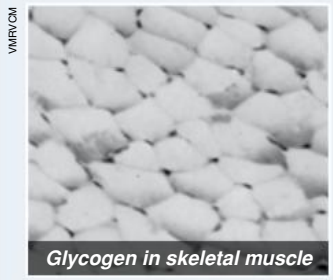
Glycogen, like starch, is a branched polysaccharide. It is chemically similar to amylopectin, being composed of α-glucose molecules, but there are more α-1,6 glycosidic links mixed with α-1,4 links. This makes it more highly branched and water-soluble than starch. Glycogen is a storage compound in animal tissues and is found mainly in liver and muscle cells (photo, right). It is readily hydrolysed by enzymes to form glucose.

Chitin

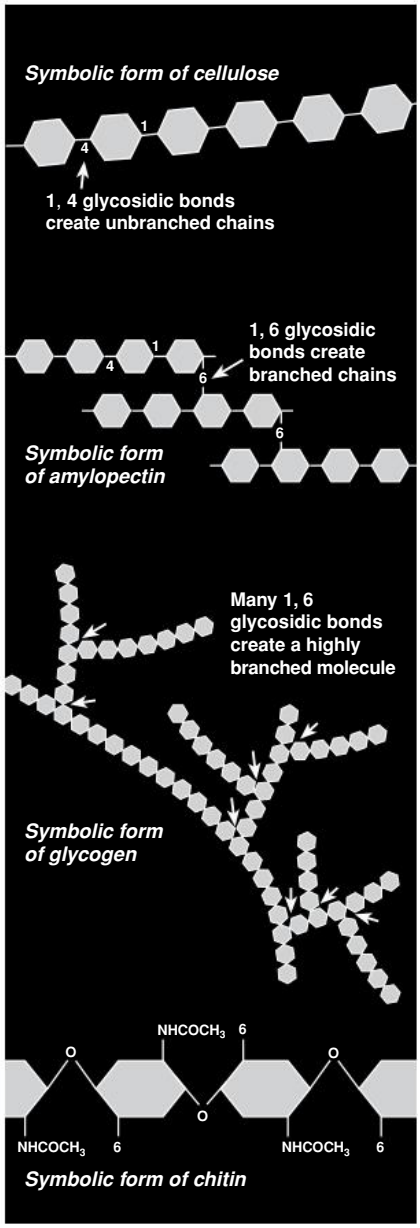
Chitin is a tough modified polysaccharide made up of chains of β-glucose molecules. It is chemically similar to cellulose but each glucose has an amine group (–NH_2) attached. After cellulose, chitin is the second most abundant carbohydrate. It is found in the cell walls of fungi and is the main component of the exoskeleton of insects (right) and other arthropods.











1. Why are polysaccharides such a good source of energy? _____

2. Discuss the structural differences between the polysaccharides starch and glycogen, explaining how the differences in structure contribute to the functional properties of the molecule:

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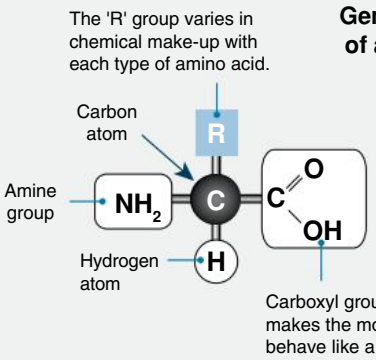
7 Amino Acids

Key Idea: Amino acids are the basic unit of proteins. They are joined by condensation reactions to form polypeptides. Proteins are made up of one or more polypeptides.

Amino acids are the basic units from which proteins are made. Twenty amino acids commonly occur in proteins and they can be linked in many different ways by peptide bonds to form a huge variety of polypeptides. Peptide bonds are formed by **condensation reactions** between amino acids. The reverse reaction, which breaks up peptide chains, uses water and is called **hydrolysis**.

Structure of Amino Acids

There are over 150 amino acids found in cells, but only 20 occur commonly in proteins. The remaining, non-protein amino acids have roles as intermediates in metabolic reactions, or as neurotransmitters and hormones. All amino acids have a common structure (see right). The only difference between the different types lies with the 'R' group in the general formula. This group is variable, which means that it is different in each kind of amino acid.



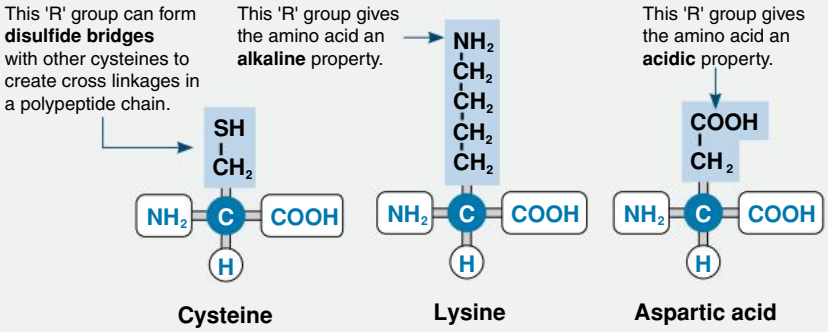
General structure of an amino acid



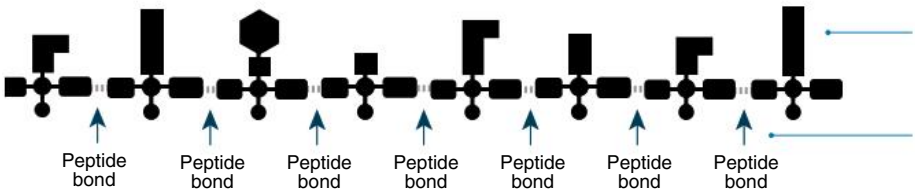
Example of an amino acid shown as a space filling model: cysteine.

Properties of Amino Acids

Three examples of amino acids with different chemical properties are shown right, with their 'R' groups outlined. The 'R' groups can have quite diverse chemical properties.



A Polypeptide Chain



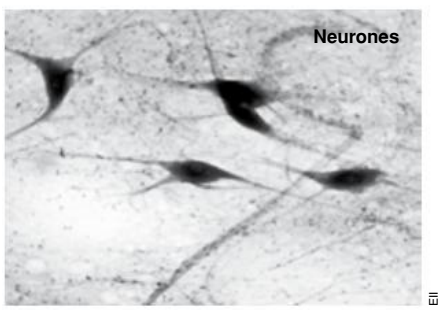
The order of amino acids in a protein is directed by the order of nucleotides in DNA and mRNA.

Peptide bonds link amino acids together in long polymers called polypeptide chains. These may form part or all of a protein

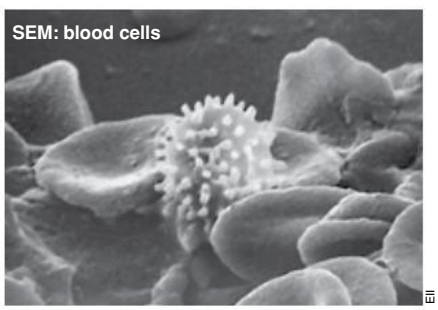
The amino acids are linked together by peptide bonds to form long chains of up to several hundred amino acids (called polypeptide chains). These chains may be functional units (complete by themselves) or they may need to be joined to other polypeptide chains before they can carry out their function. In humans, not all amino acids can be manufactured by our body: 10 must be taken in with our diet (eight in adults). These are the 'essential amino acids'. They are indicated by the symbol ♦ on the right. Those indicated with as asterisk (*) are also required by infants.

Amino acids occurring in proteins

Alanine	Glutamine	Leucine ♦	Serine
Arginine *	Glutamic acid	Lysine ♦	Threonine ♦
Asparagine	Glycine	Methionine ♦	Tryptophan ♦
Aspartic acid	Histidine *	Phenylalanine ♦	Tyrosine
Cysteine	Isoleucine ♦	Proline	Valine ♦



Several amino acids act as neurotransmitters in the central nervous system. Glutamic acid and GABA (gamma amino butyric acid) are the most common. Others, such as glycine, are restricted to the spinal cord.



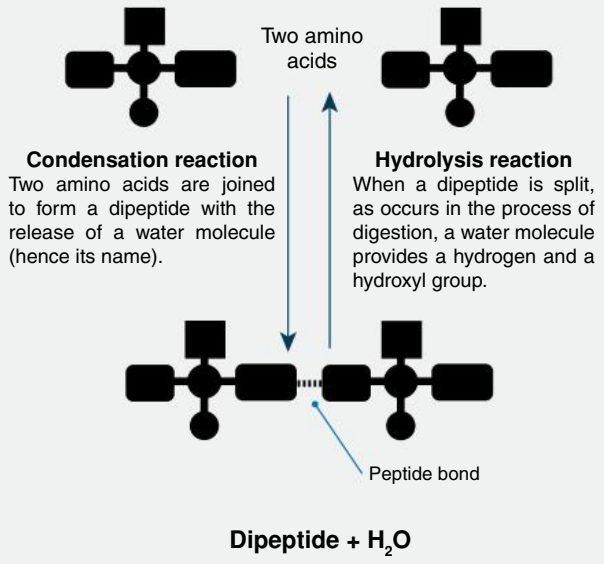
Amino acids tend to stabilise the pH of solutions (e.g. blood and tissue fluid) because they remove excess H^+ or OH^- ions. They retain this buffer capacity even when incorporated into peptides and proteins.



Amino acids are widely available as dietary supplements. Lysine is sold as relief for *Herpes* infections and glucosamine supplements are used for alleviating the symptoms of arthritis and other joint disorders.

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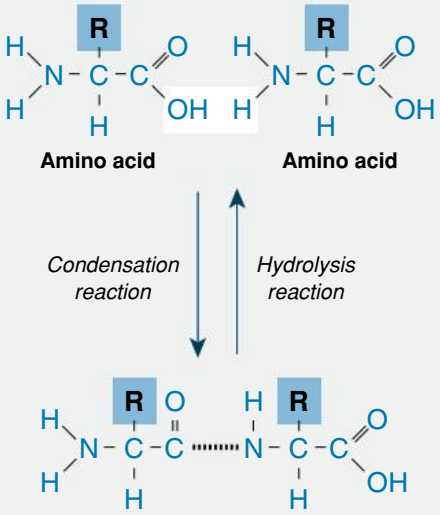
Condensation and Hydrolysis Reactions



Condensation reaction
Two amino acids are joined to form a dipeptide with the release of a water molecule (hence its name).

Hydrolysis reaction
When a dipeptide is split, as occurs in the process of digestion, a water molecule provides a hydrogen and a hydroxyl group.

Dipeptide + H_2O



Amino acid **Amino acid**

Condensation reaction

Hydrolysis reaction

Dipeptide + H_2O

1. What makes each of the 20 amino acids found in proteins unique? _____

2. Describe the process that determines the sequence in which amino acids are linked together to form polypeptide chains:

3. How does the chemical structure of amino acids enable them to act as buffers in biological tissues? _____

4. Giving examples, explain what is meant by an **essential amino acid**: _____

5. Explain how amino acids are joined together and broken down: _____

6. Discuss the various biological roles of amino acids: _____

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8 Proteins

Key Idea: Interactions between amino acid R groups direct a polypeptide chain to fold into its functional shape.

The precise folding of a protein into its **tertiary structure** creates a three dimensional arrangement of the active 'R' groups. The tertiary structure gives a protein its unique chemical properties. If this is lost (through **denaturation**), the protein is usually unable to carry out its biological function. Proteins can be classified on the basis of structure (e.g. globular vs fibrous) or function. The entire collection of proteins in a particular cell type is termed the **cellular proteome**. An organism's proteome is larger than its genome because there are more proteins than genes. This is because of alternative splicing of genes and because many proteins are modified after they are translated.

Primary Structure - 1° (amino acid sequence)

Strings of hundreds of amino acids link together with peptide bonds to form molecules called polypeptide chains. There are 20 different kinds of amino acids that can be linked together in a vast number of different combinations. This sequence is called the primary structure. It is the arrangement of attraction and repulsion points in the amino acid chain that determines the higher levels of organisation in the protein and its biological function.

Secondary Structure - 2° (α-helix or β pleated sheet)

Polypeptides become folded in various ways, referred to as the secondary (2°) structure. The most common types of 2° structures are a coiled α-helix and a β-pleated sheet. Secondary structures are maintained with hydrogen bonds between neighbouring CO and NH groups. H-bonds, although individually weak, provide considerable strength when there are a large number of them. The example, right, shows the two main types of secondary structure. In both, the **'R' side groups** (not shown) project out from the structure. Most globular proteins contain regions of α-helices together with β-sheets. Keratin (a fibrous protein) is composed almost entirely of α-helices. Fibroin (silk protein), is another fibrous protein, almost entirely in β-sheet form.

Tertiary Structure - 3° (folding)

Every protein has a precise structure formed by the folding of the secondary structure into a complex shape called the tertiary structure. The protein folds up because various points on the secondary structure are attracted to one another. The strongest links are caused by bonding between neighbouring cysteine amino acids which form disulfide bridges. Other interactions that are involved in folding include weak ionic and hydrogen bonds as well as hydrophobic interactions.

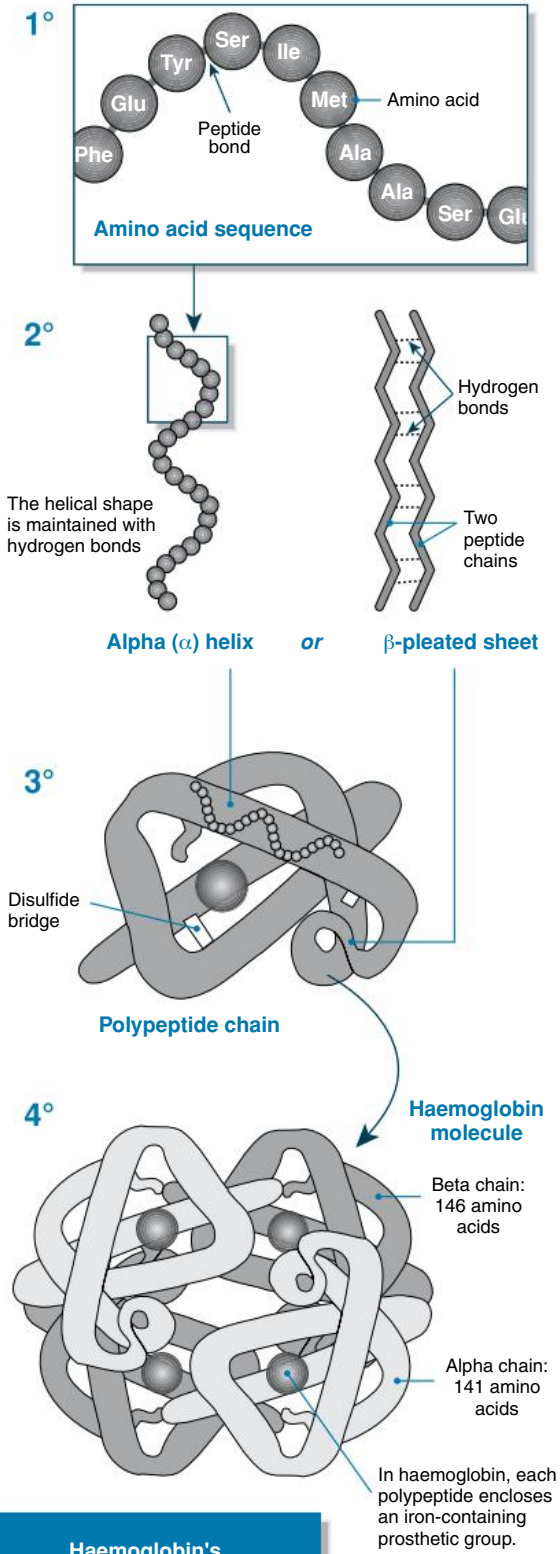
Quaternary Structure - 4°

Some proteins (such as enzymes) are complete and functional with a tertiary structure only. However, many complex proteins exist as aggregations of polypeptide chains. The arrangement of the polypeptide chains into a functional protein is termed the **quaternary structure**. The example (right) shows a molecule of haemoglobin, a globular protein composed of 4 polypeptide sub-units joined together; two identical ***beta chains*** and two identical ***alpha chains***. Each has a haem (iron containing) group at the centre of the chain, which binds oxygen. Proteins containing non-protein material are **conjugated proteins**. The non-protein part is the **prosthetic group**.

Denaturation of Proteins

Denaturation refers to the loss of the three-dimensional structure (and usually also the biological function) of a protein. Denaturation is often, although not always, permanent. It results from an alteration of the bonds that maintain the secondary and tertiary structure of the protein, even though the sequence of amino acids remains unchanged. Agents that cause denaturation are:

- **Strong acids and alkalis:** Disrupt ionic bonds and result in coagulation of the protein. Long exposure also breaks down the primary structure of the protein.
- **Heavy metals:** May disrupt ionic bonds, form strong bonds with the carboxyl groups of the R groups, and reduce protein charge. The general effect is to cause the precipitation of the protein.
- **Heat and radiation (e.g. UV):** Cause disruption of the bonds in the protein through increased energy provided to the atoms.
- **Detergents and solvents:** Form bonds with the non-polar groups in the protein, thereby disrupting hydrogen bonding.



Haemoglobin's chemical formula:

$C_{3032}H_{4816}O_{872}N_{780}S_8Fe_4$

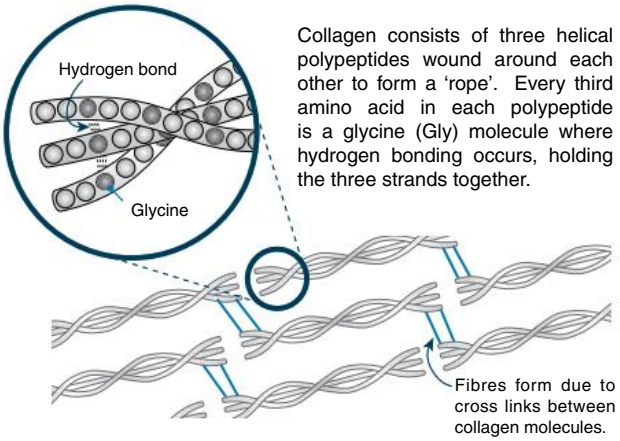
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Structural Classification of Proteins

Fibrous Proteins

Properties
- Water insoluble
- Very tough physically; may be supple or stretchy
- Parallel polypeptide chains in long fibres or sheets

Function
- Structural role in cells and organisms e.g. *collagen found in connective tissue, cartilage, bones, tendons, and blood vessel walls.*
- Contractile e.g. *myosin, actin*

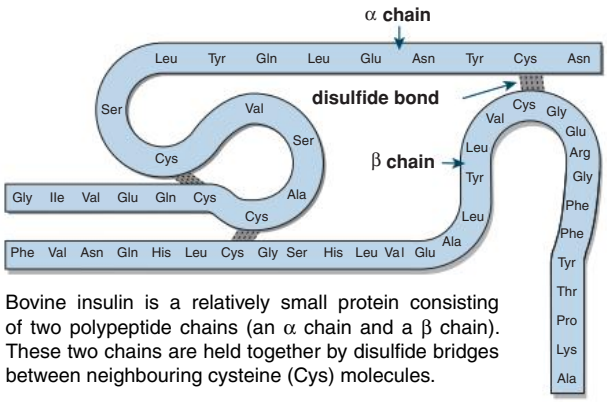


Collagen consists of three helical polypeptides wound around each other to form a 'rope'. Every third amino acid in each polypeptide is a glycine (Gly) molecule where hydrogen bonding occurs, holding the three strands together.

Globular Proteins

Properties
- Easily water soluble
- Tertiary structure critical to function
- Polypeptide chains folded into a spherical shape

Function
- Catalytic *e.g. enzymes*
- Regulatory *e.g. hormones (insulin)*
- Transport *e.g. haemoglobin*
- Protective *e.g. antibodies*



Bovine insulin is a relatively small protein consisting of two polypeptide chains (an α chain and a β chain). These two chains are held together by disulfide bridges between neighbouring cysteine (Cys) molecules.

1. Giving examples, explain the functional roles of proteins in each of the following:

   (a) Structural tissues of the body: _____

   _____

   (b) Regulating body processes: _____

   _____

   (c) Contractile elements: _____

   _____

   (d) Immunological response to pathogens: _____

   _____

   (e) Transporting molecules within cells and in the bloodstream: _____

   _____

   (f) Catalysing metabolic reactions in cells: _____

   _____

2. Explain denaturation and its effect on protein function: _____

   _____

   _____

3. Why are fibrous proteins important as structural molecules both within cells and extracellularly? _____

   _____

   _____

4. Suggest why many globular proteins, in contrast to fibrous proteins, have a catalytic or regulatory role:

   _____

   _____

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9 Modification of Proteins

Key Idea: The modification of proteins allows the cell to specify their use and final destination.

Proteins may be modified after they have been produced by **post translational modification**. Two important modifications are to add carbohydrates or lipids to the protein. **Glycoproteins** are formed by adding carbohydrates to proteins once they pass into the rough endoplasmic reticulum. The carbohydrates may help position and orientate the glycoprotein in the membrane, guide a protein to its final destination, or help in cell-to-cell recognition and cell signalling. Other proteins may have fatty acids added to them to form **lipoproteins**. These modified proteins transport lipids in the plasma between various organs in the body (e.g. gut, liver, and adipose tissue). Other common post-translational modifications include degradation, cleavage, and phosphorylation (below).

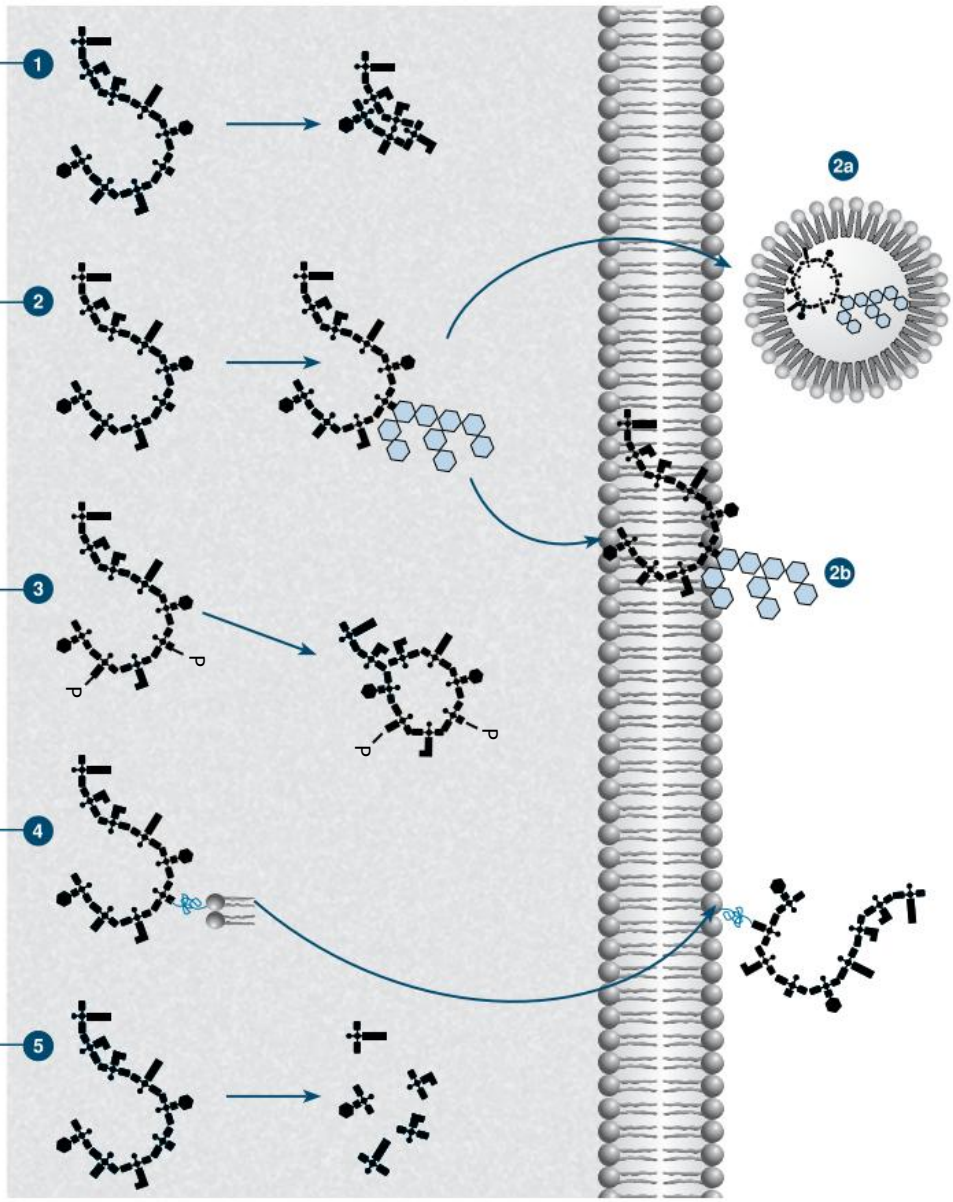
Cleaving: Polypeptide chains may be cleaved to give smaller chains, which then fold or join to give the functional protein. An example is human insulin which is transcribed as one long polypeptide chain before being cleaved in two places to form two shorter chains that form the functional protein.

Glycosylation (adding carbohydrate groups): This is used to add an ID tag to the protein that will allow the cell to recognise its use and where it is to be transported (2a). The resulting glycoprotein may be used in the cell membrane or secreted. The carbohydrate tag may help position the glycoprotein within the membrane (2b).

Phosphorylation (the addition of phosphate groups). This may contribute to the protein's three dimensional structure, or help with cell signalling.

Lipid attachment: Proteins may have lipids attached to them which anchor the protein to the plasma membrane.

Degradation: Some polypeptide chains may be tagged for degradation when they are no longer useful and their amino acids reused in the formation of other proteins.



1. (a) Describe some of the modifications that polypeptide chains undergo before becoming functional proteins:

_____

_____

_____

(b) Why are these changes necessary? _____

_____

_____

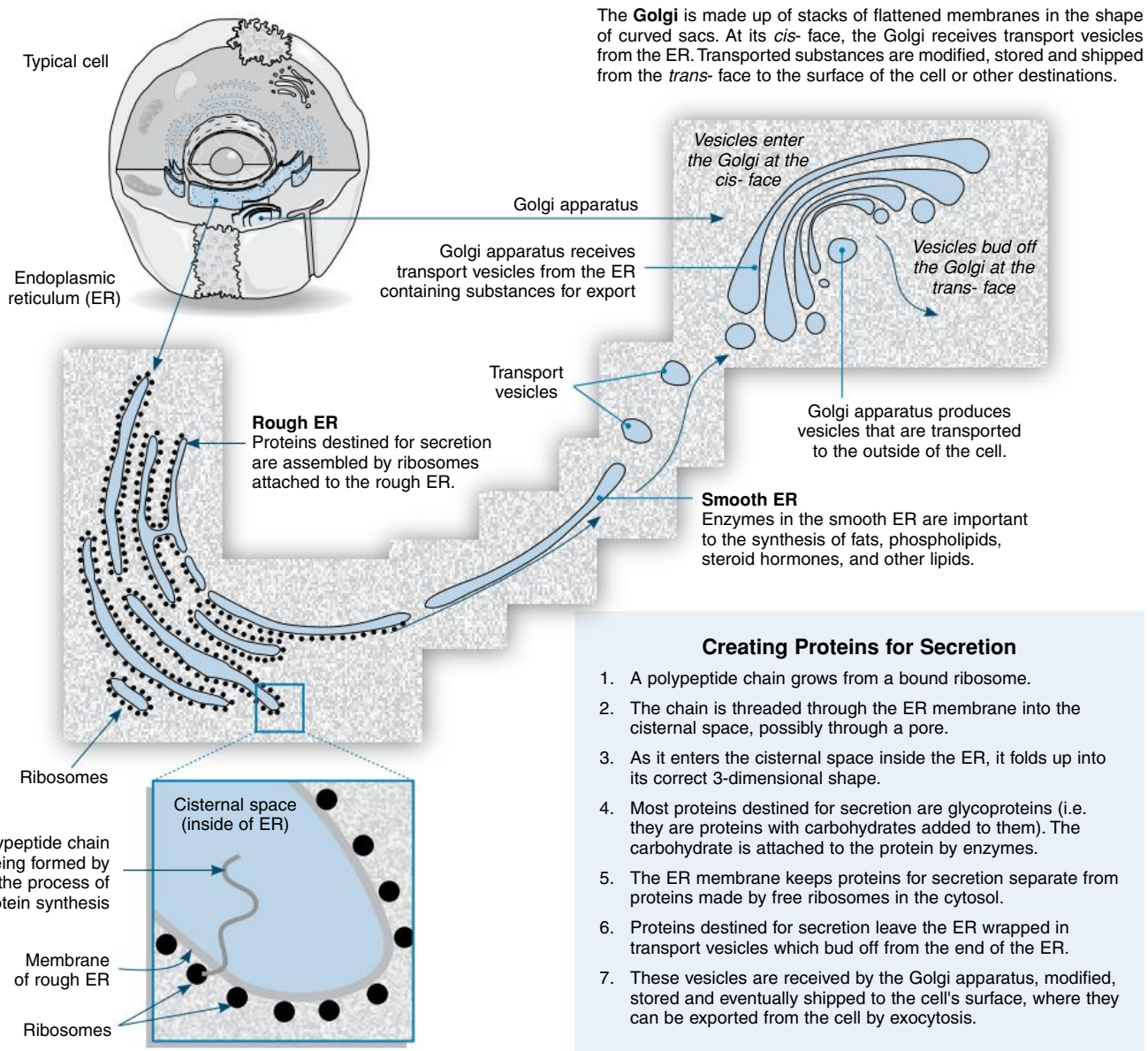
2. Why might the orientation of a protein in the plasma membrane be important? _____

_____

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10 Packaging Macromolecules

Key Idea: The synthesis, packaging and movement of macromolecules inside the cell involves many membrane-bound organelles. These organelles provide compartments where the enzyme systems involved can be isolated.



The **Golgi** is made up of stacks of flattened membranes in the shape of curved sacs. At its *cis-* face, the Golgi receives transport vesicles from the ER. Transported substances are modified, stored and shipped from the *trans-* face to the surface of the cell or other destinations.

Creating Proteins for Secretion

1. A polypeptide chain grows from a bound ribosome.
2. The chain is threaded through the ER membrane into the cisternal space, possibly through a pore.
3. As it enters the cisternal space inside the ER, it folds up into its correct 3-dimensional shape.
4. Most proteins destined for secretion are glycoproteins (i.e. they are proteins with carbohydrates added to them). The carbohydrate is attached to the protein by enzymes.
5. The ER membrane keeps proteins for secretion separate from proteins made by free ribosomes in the cytosol.
6. Proteins destined for secretion leave the ER wrapped in transport vesicles which bud off from the end of the ER.
7. These vesicles are received by the Golgi apparatus, modified, stored and eventually shipped to the cell's surface, where they can be exported from the cell by exocytosis.

1. Using examples, explain what is meant by a macromolecule: _____

_____

2. Why are polypeptides requiring transport synthesised by membrane-bound (rather than free) ribosomes?

_____

_____

3. Why are most proteins destined for secretion from the cell glycoproteins? _____

_____

4. Briefly describe the roles of the following organelles in the production of macromolecules:

   (a) Rough ER: _____

   (b) Smooth ER: _____

   (c) Golgi apparatus: _____

   (d) Transport vesicles: _____

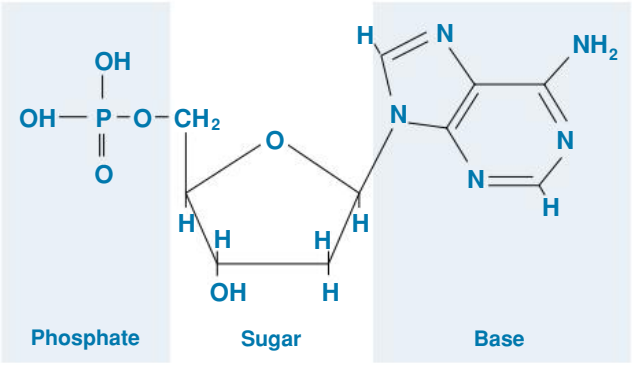
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11 Nucleic Acids

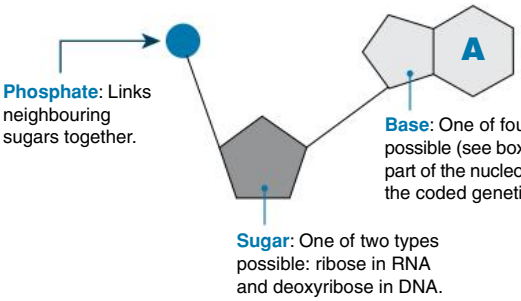
Key Idea: Nucleotides are the building blocks of DNA and RNA. Nucleic acids are long chains of nucleotides which store and transmit genetic information.

A nucleotide has three components: a base, a sugar, and a phosphate group. They are the building blocks of nucleic acids (DNA and RNA), which are involved in the transmission of inherited information. Nucleic acids have the capacity to store the information that controls cellular activity. The central nucleic acid is called deoxyribonucleic acid (DNA). Ribonucleic acids (RNA) are involved in the 'reading' of the DNA information. All nucleic acids are made up of nucleotides linked together to form chains or strands. The strands vary in the sequence of the bases found on each nucleotide. It is this sequence which provides the 'genetic code' for the cell.

Chemical Structure of a Nucleotide

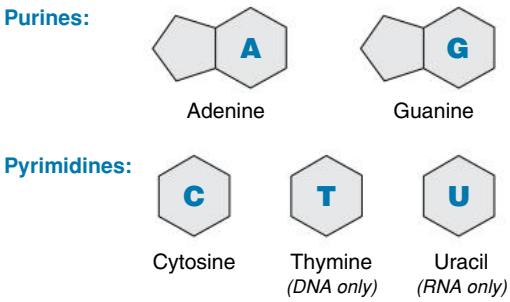


Symbolic Form of a Nucleotide



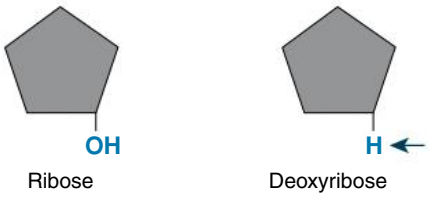
Nucleotides are the building blocks of DNA. Their precise sequence in a DNA molecule provides the genetic instructions for the organism to which it governs. Accidental changes in nucleotide sequences are a cause of mutations, usually harming the organism, but occasionally providing benefits.

Bases



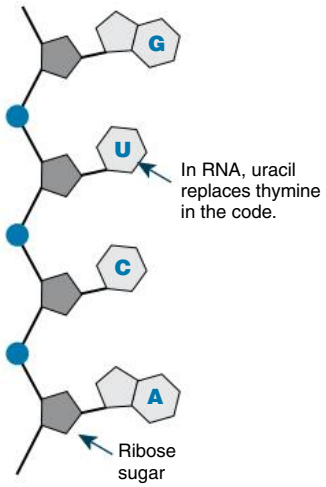
The two-ringed bases above are purines and make up the longer bases. The single-ringed bases are pyrimidines. Although only one of four kinds of base can be used in a nucleotide, uracil is found only in RNA, replacing thymine. DNA contains: A, T, G, and C, while RNA contains A, U, G, and C.

Sugars



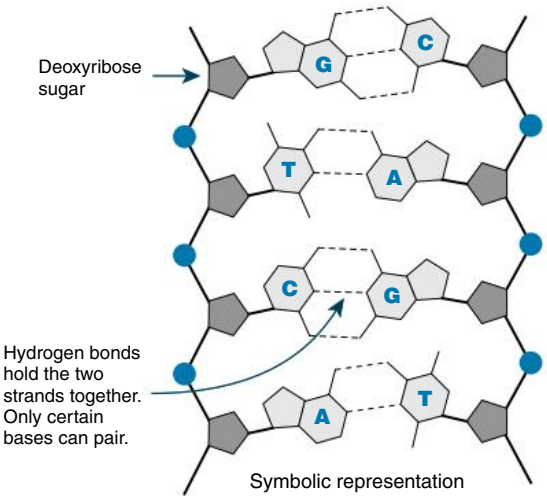
Deoxyribose sugar is found only in DNA. It differs from **ribose** sugar, found in RNA, by the lack of a single oxygen atom (arrowed).

RNA Molecule

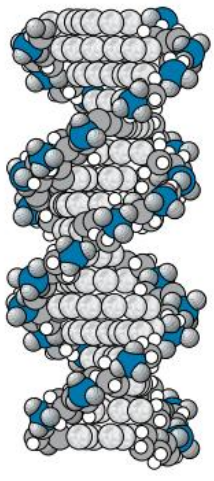


Ribonucleic acid (RNA) comprises a single strand of nucleotides linked together.

DNA Molecule



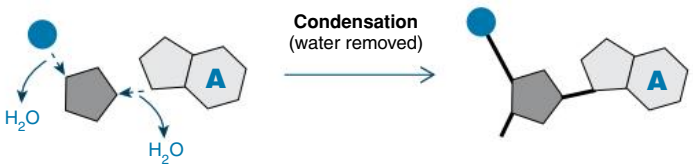
DNA Molecule



Deoxyribonucleic acid (DNA) comprises a double strand of nucleotides linked together. It is shown unwound in the symbolic representation (left). The DNA molecule takes on a twisted, double helix shape as shown in the space filling model on the right.

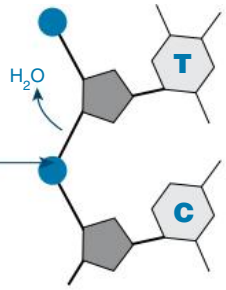
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Formation of a nucleotide



A nucleotide is formed when phosphoric acid and a base are chemically bonded to a sugar molecule. In both cases, water is given off, and they are therefore condensation reactions. In the reverse reaction, a nucleotide is broken apart by the addition of water **(hydrolysis)**.

Formation of a dinucleotide

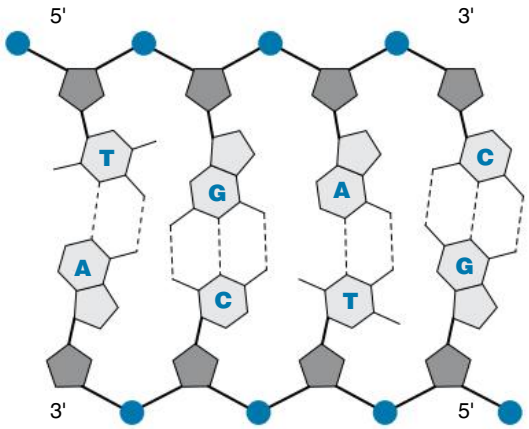


Two nucleotides are linked together by a condensation reaction between the phosphate of one nucleotide and the sugar of another.

Double-Stranded DNA

The double-helix structure of DNA is like a ladder twisted into a corkscrew shape around its longitudinal axis. It is 'unwound' here to show the relationships between the bases.

- The DNA backbone is made up from alternating phosphate and sugar molecules, giving the DNA molecule an asymmetrical structure.
- The asymmetrical structure gives a DNA strand a **direction**. Each strand runs in the opposite direction to the other.
- The ends of a DNA strand are labelled the 5' (five prime) and 3' (three prime) ends. The **5'** end has a terminal phosphate group (off carbon 5), the **3'** end has a terminal hydroxyl group (off carbon 3).
- The way the pairs of bases come together to form hydrogen bonds is determined by the number of bonds they can form and the configuration of the bases.



1. The diagram above depicts a double-stranded DNA molecule. Label the following parts on the diagram:
   (a) **Sugar** (deoxyribose)
   (b) **Phosphate**
   (c) **Hydrogen bonds** (between bases)
   (d) **Purine** bases
   (e) **Pyrimidine** bases

2. (a) Explain the **base-pairing rule** that applies in double-stranded DNA: _____

   _____

   (b) How is the base-pairing rule for mRNA different? _____

   (c) What is the purpose of the hydrogen bonds in double-stranded DNA? _____

   _____

3. Describe the functional role of nucleotides: _____

   _____

4. (a) Why do the DNA strands have an asymmetrical structure? _____

   _____

   (b) What are the differences between the 5' and 3' ends of a DNA strand? _____

   _____

5. Complete the following table summarising the differences between DNA and RNA molecules:

	DNA	RNA
Sugar present		
Bases present		
Number of strands		
Relative length		

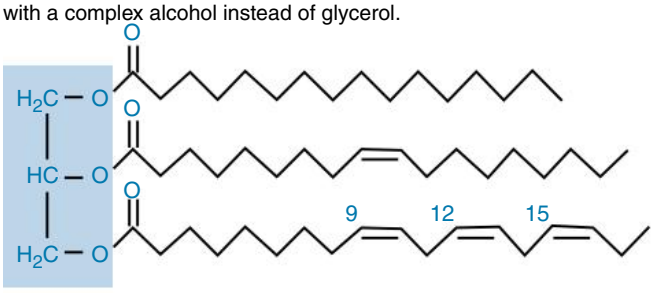
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12 Lipids

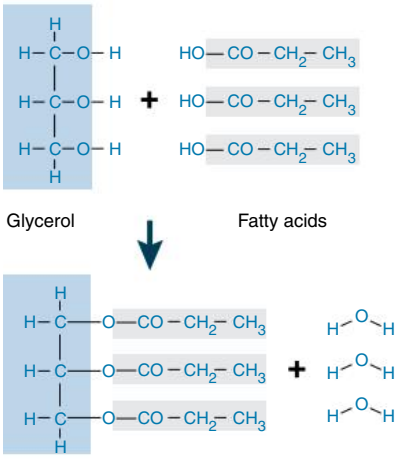
Key Idea: Lipids are molecules that serve as a source and storage of energy, and as structural components of cells. Lipids are a group of organic compounds with an oily, greasy, or waxy consistency. They are relatively insoluble in water and tend to be water-repelling. Lipids are important biological fuels, some are hormones, and some serve as structural components in plasma membranes. Proteins and carbohydrates may be converted into fats by enzymes and stored within cells of adipose tissue. During times of plenty, this store is increased, to be used when food is short.

Neutral Fats and Oils

The most abundant lipids in living things are neutral fats. They make up the fats and oils found in plants and animals. Fats are an economical way to store fuel reserves, since they yield more than twice as much energy as the same quantity of carbohydrate. Neutral fats are composed of a glycerol molecule attached to one (monoglyceride), two (diglyceride) or three (triglyceride) fatty acids. The fatty acid chains may be saturated or unsaturated (see below). Waxes are similar in structure to fats and oils, but they are formed with a complex alcohol instead of glycerol.



Triglyceride: an example of a neutral fat



Condensation

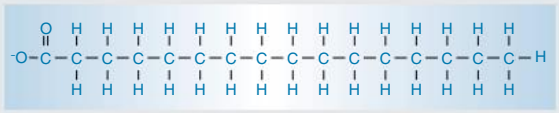
Triglycerides form when glycerol bonds with three fatty acids. Glycerol is an alcohol containing three carbons. Each of these carbons is bonded to a hydroxyl (-OH) group.

When glycerol bonds with the fatty acid, an ester bond is formed and water is released. Three separate condensation reactions are involved in producing a triglyceride.

Saturated and Unsaturated Fatty Acids

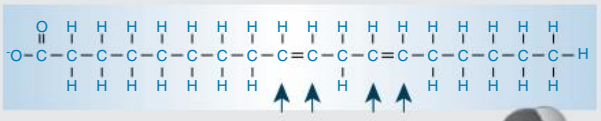
Fatty acids are a major component of neutral fats and phospholipids. About 30 different kinds are found in animal lipids. Saturated fatty acids contain the maximum number of hydrogen atoms. Unsaturated fatty acids contain some carbon atoms that are double-bonded with each other and are not fully saturated with hydrogens.

Lipids containing a high proportion of saturated fatty acids tend to be solids at room temperature (e.g. butter). Lipids with a high proportion of unsaturated fatty acids are oils and tend to be liquid at room temperature. This is because the unsaturation causes kinks in the straight chains so that the fatty acids do not pack together. Regardless of their degree of saturation, fatty acids yield a large amount of energy when oxidised.

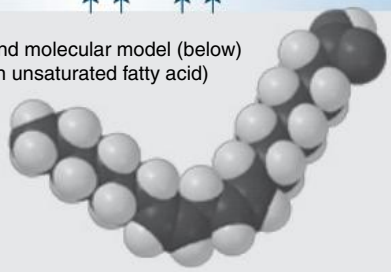


Formula (above) and molecular model (below) for **palmitic acid** (a saturated fatty acid)





Formula (above) and molecular model (below) for linoleic acid (an unsaturated fatty acid)



1. (a) Distinguish between saturated and unsaturated fatty acids: _____

_____

(b) Relate the properties of a neutral fat or phospholipid to the type of fatty acid present: _____

_____

_____

_____

2. Explain how neutral fats can provide an animal with:

(a) Energy: _____

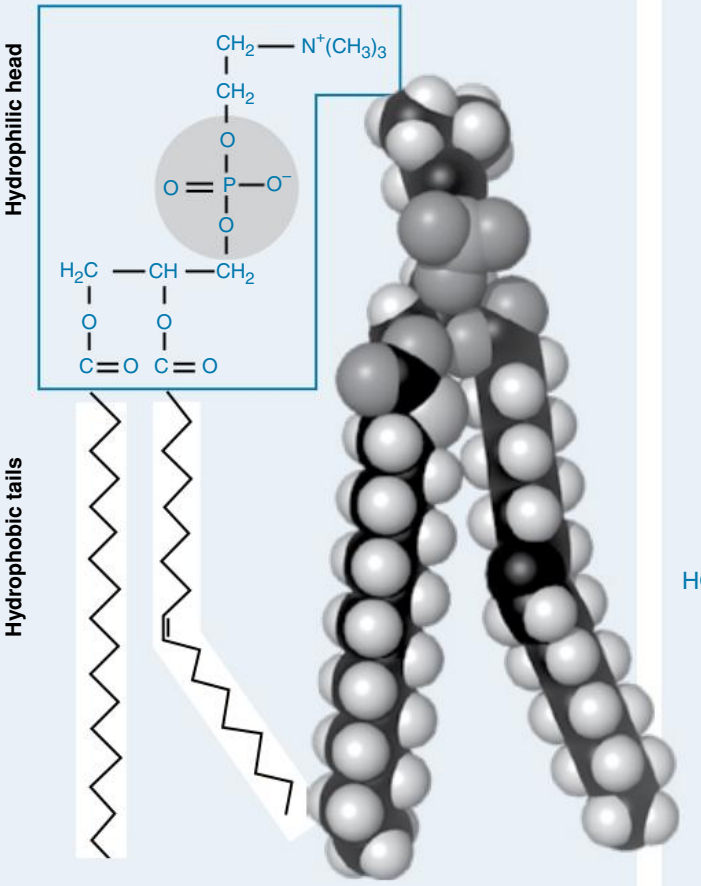
(b) Water: _____

(c) Insulation: _____

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Phospholipids

Phospholipids are the main component of cellular membranes. They consist of a glycerol attached to two fatty acid chains and a phosphate (PO_4^{3-}) group. The phosphate end of the molecule is attracted to water (it is hydrophilic) while the fatty acid end is repelled (hydrophobic). The hydrophobic ends turn inwards in the membrane to form a **phospholipid bilayer.**

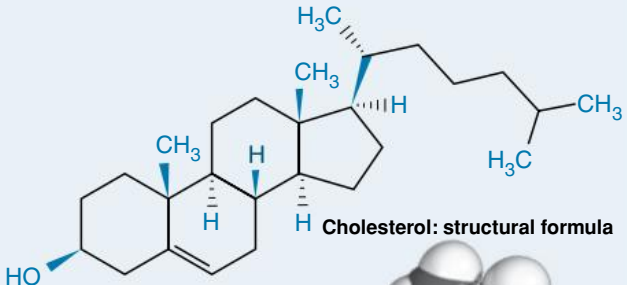


Steroids and Cholesterol

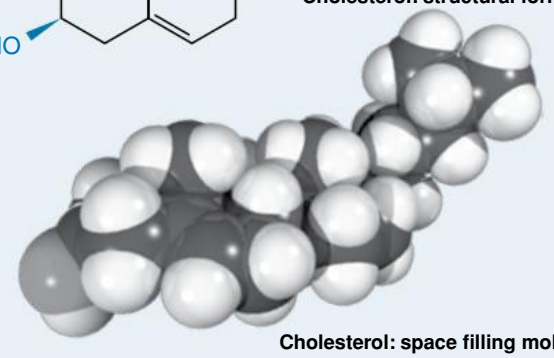
Although steroids are classified as lipids, their structure is quite different to that of other lipids. Steroids have a basic structure of three rings made of 6 carbon atoms each and a fourth ring containing 5 carbon atoms. Examples of steroids include the male and female sex hormones (testosterone and oestrogen), and the hormones cortisol and aldosterone.

Cholesterol, while not a steroid itself, is a sterol lipid and is a precursor to several steroid hormones. It is present in the plasma membrane, where it regulates membrane fluidity by preventing the phospholipids packing too closely together.

Like phospholipids, cholesterol is **amphipathic**. The hydroxyl (-OH) group on cholesterol interacts with the polar head groups of the membrane phospholipids, while the steroid ring and hydrocarbon chain tuck into the hydrophobic portion of the membrane. This helps to stabilise the outer surface of the membrane and reduce its permeability to small water-soluble molecules.



Cholesterol: structural formula



Cholesterol: space filling molecule

3. What is the key **chemical** difference between a phospholipid and a triglyceride? _____

_____

_____

4. Why are saturated fats (e.g. lard) solid at room temperature? _____

_____

5. (a) Relate the structure of phospholipids to their chemical properties and their functional role in cellular membranes:

_____

_____

(b) Suggest how the cell membrane structure of an Arctic fish might differ from that of tropical fish species:

_____

_____

6. How does the structure of cholesterol enable it to perform structural and functional roles within membranes?

_____

_____

_____

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13 Biochemical Tests

Key Idea: Biochemical tests are used to detect molecules such as lipids, proteins, and carbohydrates.

Lipids, proteins, and carbohydrates can all be detected by relatively simple tests. For tests where the presence of a substance is indicated by a colour change, **colorimetric analysis** of a known dilution series (e.g. five prepared dilutions of glucose) can be used to produce a **calibration curve**. This curve can then be used to quantify that substance in samples of unknown concentration. Simple biochemical tests are useful, but they are crude. To analyse a mix of substances, a technique such as chromatography is required. For example, a positive Benedict's test indicates the presence of reducing sugar(s), but chromatography will distinguish different sugars (e.g. fructose and glucose).

Simple Food Tests

Proteins: The Biuret Test

Reagent:	Biuret solution.
Procedure:	A sample is added to biuret solution and gently heated.
Positive result:	Solution turns from blue to lilac.

Starch: The Iodine Test

Reagent:	Iodine.
Procedure:	Iodine solution is added to the sample.
Positive result:	Blue-black staining occurs.

Lipids: The Emulsion Test

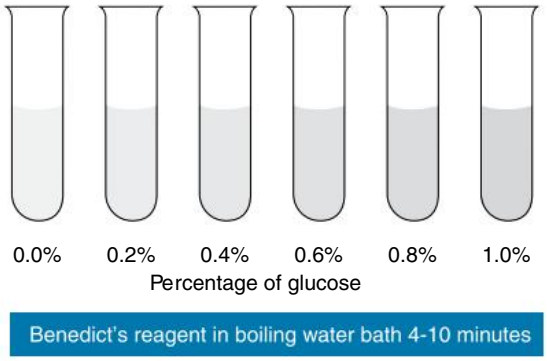
Reagent:	Ethanol.
Procedure:	The sample is shaken with ethanol. After settling, the liquid portion is distilled and mixed with water.
Positive result:	The solution turns into a cloudy-white emulsion of suspended lipid molecules.

Sugars: The Benedict's Test

Reagent:	Benedict's solution.
Procedure:	Non reducing sugars: The sample is boiled with dilute hydrochloric acid (acid hydrolysis), then cooled and neutralised. A test for reducing sugars is then performed. Reducing sugar: Benedict's solution is added, and the sample is placed in a water bath.
Positive result:	Solution turns from blue to orange to red-brown.

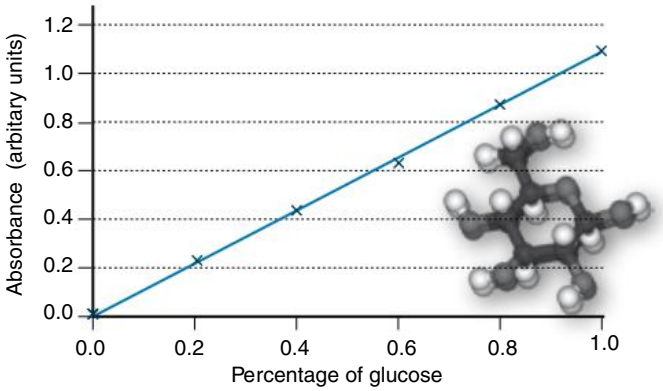
Colorimetric Analysis of Glucose

Prepare glucose standards



Produce the calibration curve

Cool and filter samples as required. Using a red filter, measure the absorbance (at 735 nm) for each of the known dilutions and use these values to produce a calibration curve for glucose.



1. Explain why lipids must be mixed in ethanol before they will form an emulsion in water: _____

2. (a) Explain how you could quantify the amount of glucose in a range of commercially available glucose drinks:

(b) Explain how you would proceed if the absorbance values you obtained for most of your 'unknowns' were outside the range of your calibration curve:

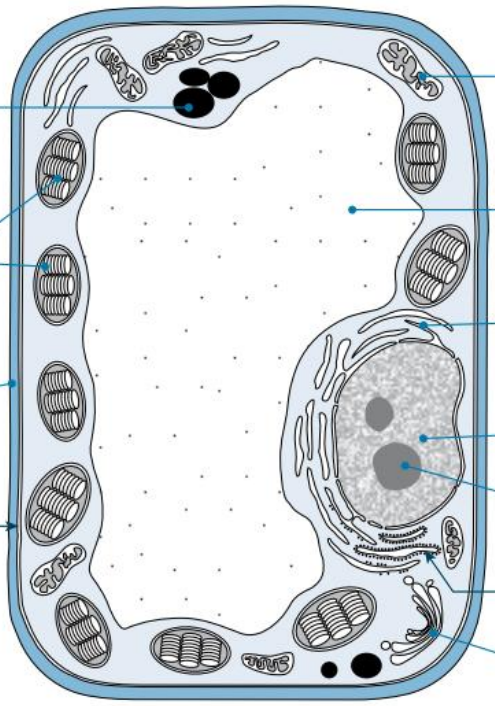
3. Explain the purpose of acid hydrolysis of a non-reducing sugar before testing with Benedict's reagent: _____

4. Explain why the emulsion of lipids, ethanol, and water appears cloudy: _____

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14 Plant and Animal Cell Organelles

Key Idea: Eukaryotic cells share many of the same structures and organelles, but some are unique to a particular cell type. Animal cells lack a cell wall, chloroplasts, and large vacuoles. Higher plant cells lack centrioles. All cells have a plasma membrane that forms the outer boundary of the cell. Many cellular organelles are also enclosed by one or two membranes. Some components of plant and animal cells are described in summary below.



Starch granule: carbohydrate stored in non-photosynthetic plastid.

Chloroplasts (2 µm x 5 µm): membranous organelles containing the green pigment chlorophyll. They contain dense stacks of membranes (grana) within a colourless stroma. They are the sites for photosynthesis.

Cell wall (0.1 µm to several µm thick): a semi-rigid structure, composed mainly of cellulose. It supports the cell and limits its volume.

Plasma membrane (3 to 10 nm thick): phospholipid bilayer with embedded proteins. Located inside the cell wall in plant cells.

Mitochondrion

Large central vacuole: usually filled with an aqueous solution of ions. Vacuoles are prominent in plants and function in storage, waste disposal, and growth. If present, vacuoles are small in animal cells.

Smooth endoplasmic reticulum (SER)

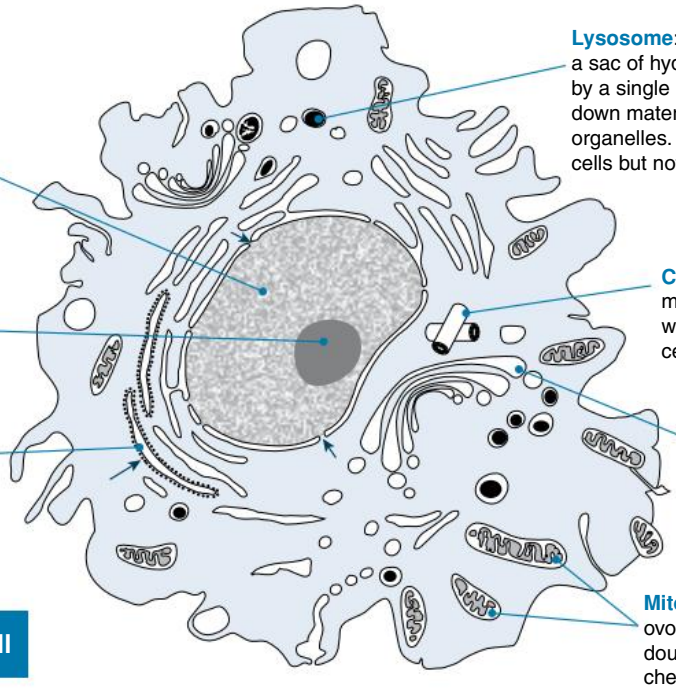
Nucleus

Nucleolus

Rough endoplasmic reticulum (RER)

Golgi apparatus

Generalised Plant Cell



Nucleus (5 µm diameter): surrounded by a double-membrane, it contains most of the cell's DNA. Pores (arrows) allow communication with the cell cytoplasm.

Nucleolus: composed of protein and nucleic acid. Involved in ribosome synthesis.

Endoplasmic reticulum (ER): a network of tubes and flattened sacs. ER may be smooth or rough (with attached ribosomes (arrow).

Lysosome: a sac of hydrolytic enzymes surrounded by a single membrane. They break down material including worn-out organelles. Probably present in plant cells but not well documented.

Centrioles (0.25 µm diameter): microtubular structures associated with nuclear division. Absent in the cells of higher plants.

Golgi apparatus: a series of flattened, membranous sacs, connected with the ER. The Golgi stores, modifies, and packages proteins.

Mitochondria (1.5 µm X 2–8 µm). ovoid structures bounded by a double membrane. Convert chemical energy into ATP.

Generalised Animal Cell

1. What two obvious features distinguish the plant and animal cells in this diagram:

   (a) _____

   (b) _____

2. Rank the following organelles according to size (largest to smallest).
   Organelles: chloroplast, mitochondrion, nucleus, centrioles

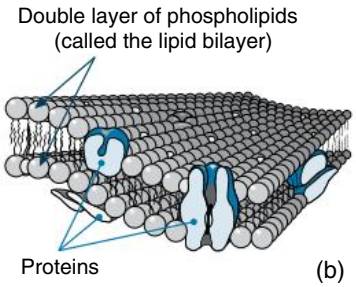
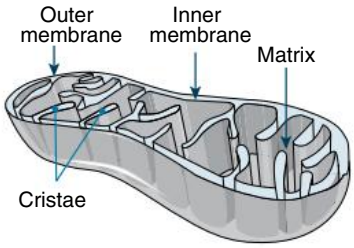
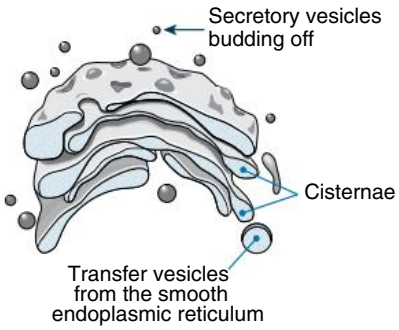
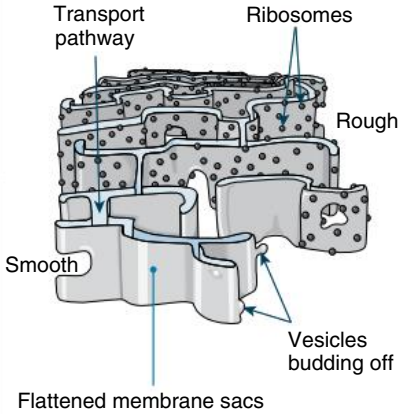
   _____

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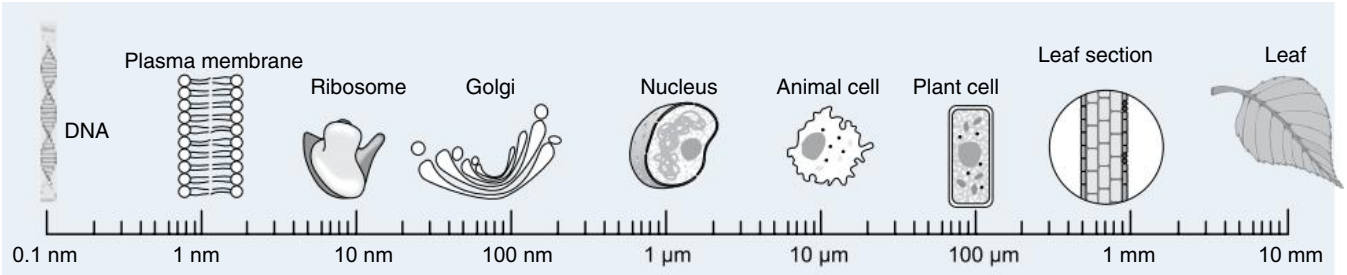
15 Review of Cell Ultrastructure

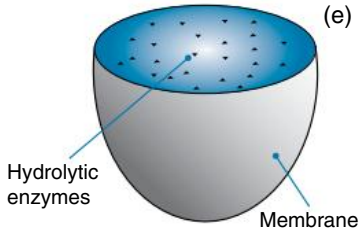
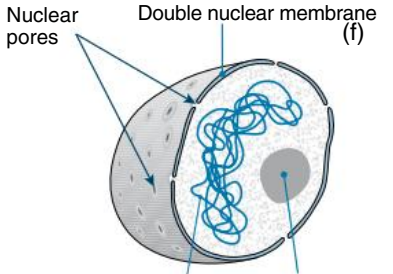
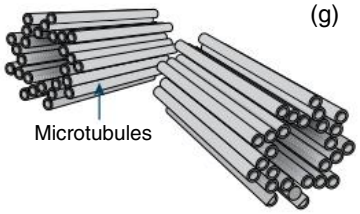
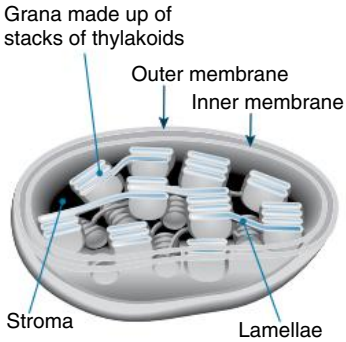
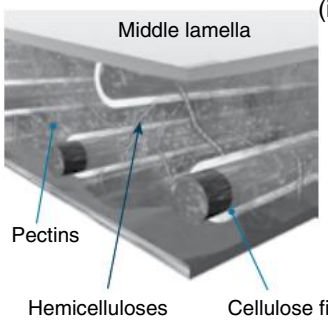
Key Idea: Eukaryotic cells contain a variety of organelles that are specialised to carry out particular tasks.

The table below provides a format to summarise information about structures and organelles of typical eukaryotic cells. Complete the table by referring to other pages in this chapter including the opposite page. The completed table will provide a concise summary of the function and location of important eukaryotic organelles. Some parts of the table are completed to help you. The log scale of measurements (next page) illustrates the relative sizes of some cellular structures.

Cell Component	Details	Present in: Plant cells	Present in: Animal cells
(a) 	Name: Plasma (cell surface) membrane Location: Surrounding the cell Function: Gives the cell shape and protection. It also regulates the movement of substances into and out of the cell.	YES	YES
(b) 	Name: Location: Function:		
(c) 	Name: Golgi apparatus Location: In cytoplasm associated with smooth ER Function:		
(d) 	Name: Smooth and rough endoplasmic reticulum Location of smooth ER: Penetrates the whole cytoplasm Location of rough ER: Function of smooth ER: Function of rough ER:		

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Cell Component	Details	Present in: Plant cells	Present in: Animal cells
(e) 	Name: Lysosome (section across) Location: Function:		
	Name: Nucleus Location: Position is variable within a cell Function:		
(g) 	Name: Centrioles Location: In cytoplasm next to nucleus Function:		
	Name: Location: Function:		
(i) 	Name: Cellulose cell wall Location: Function:		

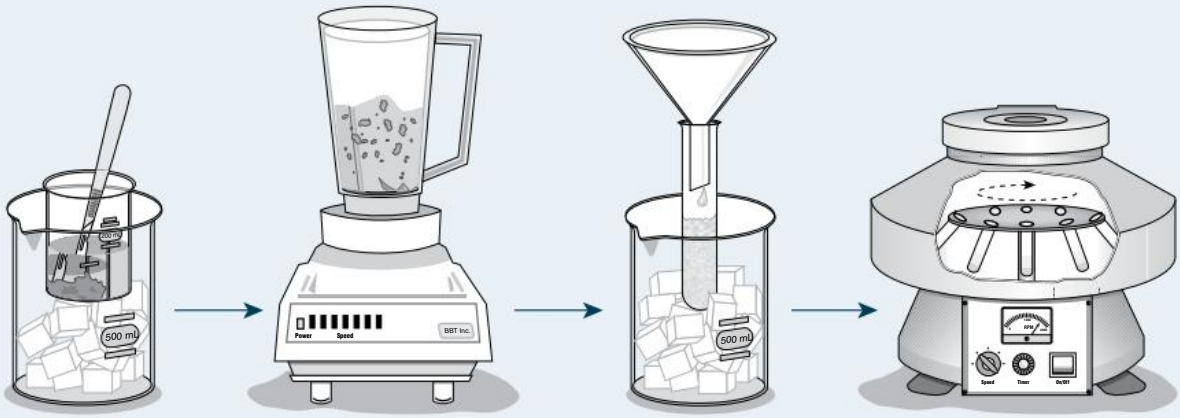
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16 Cell Fractionation

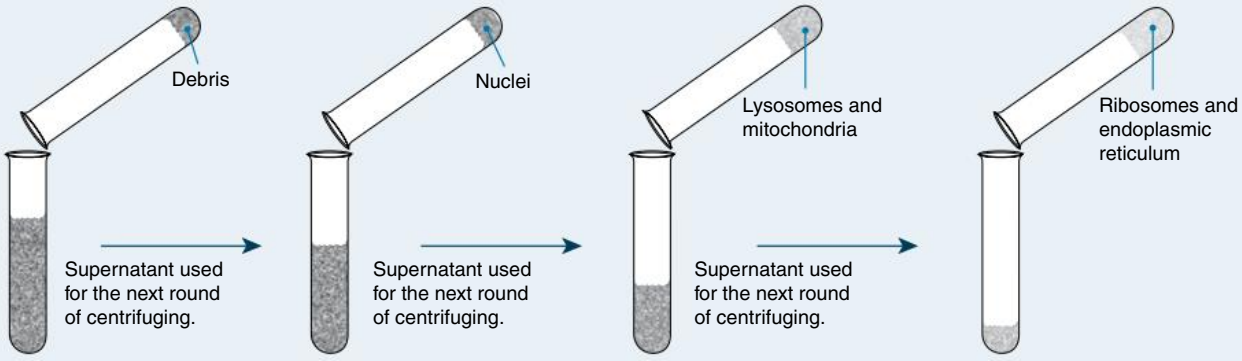
Key Idea: Cell fractionation (or differential centrifugation) is used to extract organelles from cells for study.

The aim of cell fractionation is to extract intact organelles. Samples are kept very cool so that metabolism is slowed and self digestion of the organelles is prevented. The samples are also kept in a buffered, isotonic solution so that the organelles do not change volume and the enzymes are not denatured by changes in pH.

Separating Cellular Components Using Cell Fractionation



1. The sample is chilled over ice and cut into small pieces in a cold, buffered, isotonic solution.
2. The sample is homogenised thoroughly before centrifugation. The cell organelles remain intact.
3. The homogenised suspension is filtered to remove cellular debris. It is kept cool throughout.
4. The filtrate is centrifuged at low speed to remove partially opened cells and small pieces of debris.



5. The supernatant containing the organelles is carefully decanted off.
6. The sample is centrifuged at 500-600 *g* for 5-10 minutes then decanted.
7. The sample is centrifuged at 10,000-20,000 *g* for 15-20 minutes then decanted.
8. The sample is centrifuged at 100,000 *g* for 60 minutes then decanted.

NOTE: In centrifugation, the relative centrifugal force (RCF) is expressed as 'g', where g represents the gravitational field strength.

1. Why is it possible to separate cell organelles using centrifugation? _____

_____

_____

2. Why is the sample homogenised before centrifugation? _____

_____

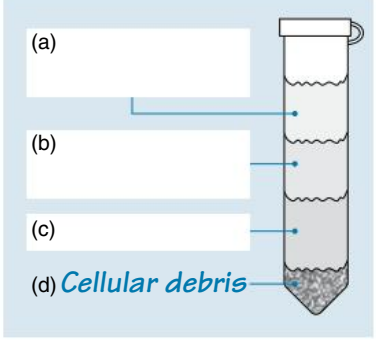
3. Explain why the sample must be kept in a solution that is:

   (a) Isotonic: _____

   (b) Cool: _____

   (c) Buffered: _____

4. **Density gradient centrifugation** (right) is another method of cell fractionation. Sucrose is added to a sample, which is then centrifuged at high speed. The organelles will form layers according to their specific densities. Using the information above, label the centrifuge tube on the right with the organelles you would find in each layer.



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17 The Role of Membranes in Cells

Key Idea: Membranes have essential roles in transport and synthesis of cellular materials, and cellular communication.

Many of the important structures and organelles in cells are made of, or are enclosed by, membranes. These include the endoplasmic reticulum, mitochondria, nucleus, Golgi body, chloroplasts, lysosomes, vesicles and the cell plasma membrane itself. All membranes within eukaryotic cells share the same basic structure as the plasma membrane that encloses the cell. They have critical roles in cells, making compartments for different reactions, controlling the entry and exit of substances, and assisting in cell-to-cell recognition and communication.

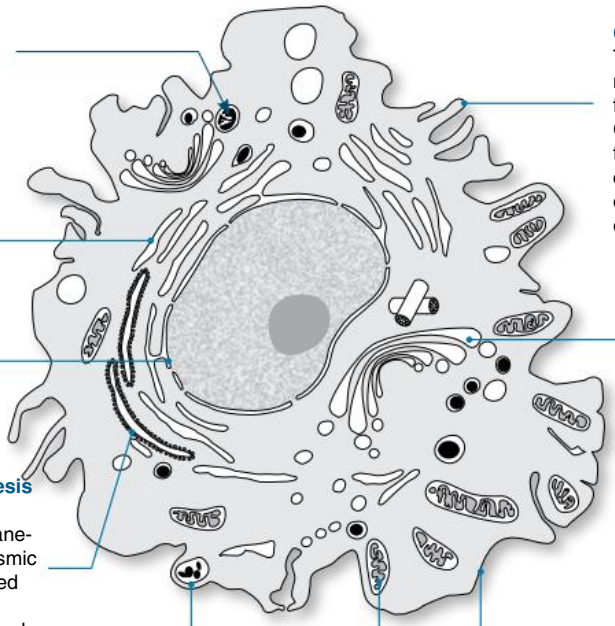
Isolation of enzymes Membrane-bound lysosomes contain enzymes for the destruction of wastes and foreign material. Peroxisomes are the site for destruction of the toxic and reactive molecule, hydrogen peroxide (formed as a result of some cellular reactions).

Role in lipid synthesis
The smooth ER is the site of lipid and steroid synthesis.

Containment of DNA
The nucleus is surrounded by a nuclear envelope of two membranes, forming a separate compartment for the cell's genetic material.

Role in protein and membrane synthesis
Some protein synthesis occurs on free ribosomes, but much occurs on membrane-bound ribosomes on the rough endoplasmic reticulum. Here, the protein is synthesised directly into the space within the ER membranes. The rough ER is also involved in membrane synthesis, growing in place by adding proteins and phospholipids.

Entry and export of substances The plasma membrane may take up fluid or solid material and form membrane-bound vesicles (or larger vacuoles) within the cell. Membrane-bound transport vesicles move substances to the inner surface of the cell where they can be exported from the cell by exocytosis.



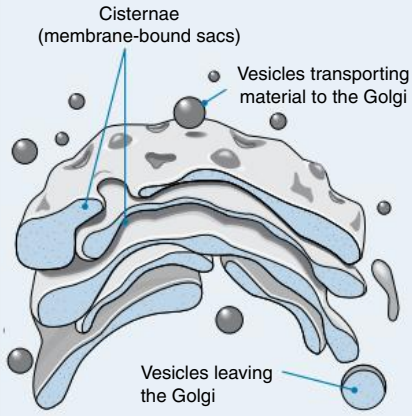
Cell communication and recognition
The proteins embedded in the membrane act as receptor molecules for hormones and neurotransmitters. Glycoproteins and glycolipids stabilise the plasma membrane and act as cell identity markers, helping cells to organise themselves into tissues, and enabling foreign cells to be recognised.

Packaging and secretion
The Golgi apparatus is a specialised membrane-bound organelle which produces lysosomes and compartmentalises the modification, packaging and secretion of substances such as proteins and hormones. See the example explained below.

Transport processes
Channel and carrier proteins are involved in selective transport across the plasma membrane. Cholesterol in the membrane can help to prevent ions or polar molecules from passing through the membrane (acting as a plug).

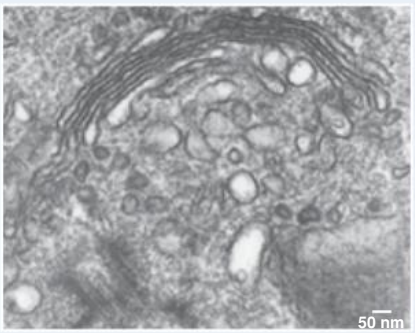
Energy transfer The reactions of cellular respiration (and photosynthesis in plants) take place in the membrane-bound energy transfer systems occurring in mitochondria and chloroplasts respectively.

Membranes Make Compartments



Membranes play an important role in separating regions within the cell (and within organelles) where particular reactions occur. Specific enzymes are therefore often located in particular organelles. The reaction rate is controlled by controlling the rate at which substrates enter the organelle and therefore the availability of the raw materials required for the reactions.

The Golgi *(diagram left and TEM right) modifies, sorts, and packages macromolecules for cell secretion. Enzymes within the cisternae modify proteins by adding carbohydrates and phosphates. To do this, the Golgi imports the substances it needs from the cytosol.*

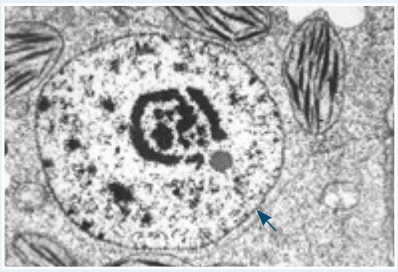


Louisa Howard-Dartmouth College PD

1. Discuss the importance of membrane systems and organelles in providing compartments within the cell:

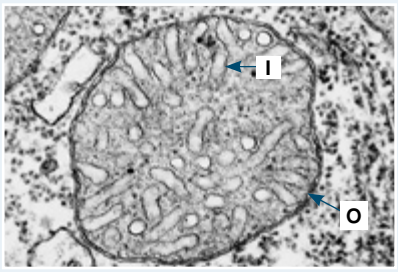
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Functional Roles of Membranes in Cells

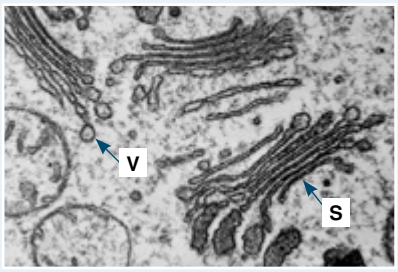


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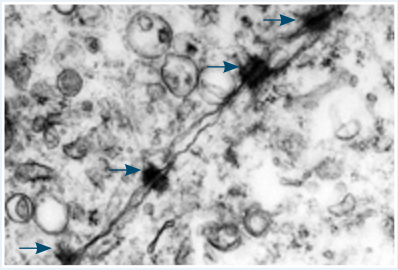
The **nuclear membrane,** which surrounds the nucleus, regulates the passage of genetic information to the cytoplasm and may also protect the DNA from damage.



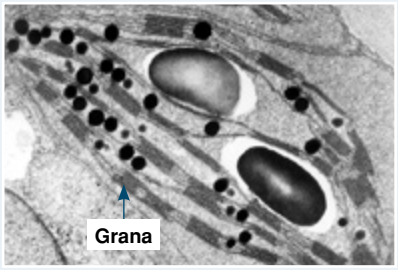
Mitochondria have an outer membrane (**O**) which controls the entry and exit of materials involved in aerobic respiration. Inner membranes (**I**) provide attachment sites for enzyme activity.



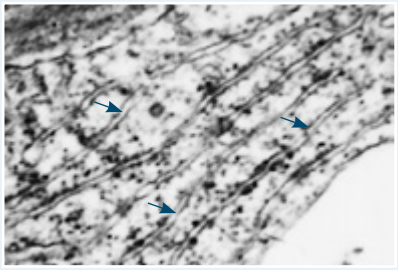
The **Golgi apparatus** comprises stacks of membrane-bound sacs (**S**). It is involved in packaging materials for transport or export from the cell as secretory vesicles (**V**).



The **plasma membrane** surrounds the cell. In this photo, intercellular junctions called **desmosomes**, which connect neighbouring cells, are indicated with arrows.



Chloroplasts are large organelles found in plant cells. The stacked membrane systems of chloroplasts (grana) trap light energy which is then used to fix carbon into 6-C sugars.



This EM shows stacks of rough endoplasmic reticulum (arrows). The membranes are studded with ribosomes, which synthesise proteins into the inter-membrane space.

Photos: WMU unless otherwise stated

2. Match each of the following organelles with the correct description of its functional role in the cell:

   chloroplast, *rough endoplasmic reticulum*, *lysosome*, *smooth endoplasmic reticulum*, *mitochondrion*, *Golgi apparatus*

   (a) Active in synthesis, sorting, and secretion of cell products: _____

   (b) Digestive organelle where macromolecules are hydrolysed: _____

   (c) Organelle where most cellular respiration occurs and most ATP is generated: _____

   (d) Active in membrane synthesis and synthesis of secretory proteins: _____

   (e) Active in lipid and hormone synthesis and secretion: _____

   (f) Photosynthetic organelle converts light energy to chemical energy stored in sugar molecules: _____

3. How is membrane surface area increased within cells and organelles? _____

   _____

4. Discuss the importance of each of the following to cellular function:

   (a) High membrane surface area: _____

   _____

   (b) Channel proteins and carrier proteins in the plasma membrane: _____

   _____

5. Non-polar (lipid-soluble) molecules diffuse more rapidly through membranes than polar (lipid-insoluble) molecules:

   (a) Explain the reason for this: _____

   _____

   (b) Discuss the implications of this to the transport of substances into the cell through the plasma membrane:

   _____

   _____

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18 Enzymes

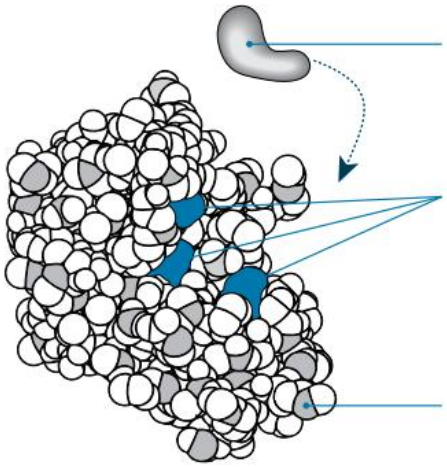
Key Idea: Enzymes are biological catalysts. The active site is critical to this functional role.

Most enzymes are proteins. They are called biological catalysts because they speed up biochemical reactions, but the enzyme itself remains unchanged. They may break down a single substrate molecule into simpler substances, or join two or more substrate molecules together. The part of the enzyme into which the substrate binds and undergoes reaction is the active site, which is formed by the folding of the enzyme's amino acid sequence.

Enzyme Structure

The model on the right illustrates the enzyme *Ribonuclease S*, which breaks up RNA molecules. It is a typical enzyme, being a globular protein and composed of up to several hundred atoms. The darkly shaded areas are part of the **active site** and make up the **cleft**; the region into which the substrate molecule(s) are drawn.

The correct positioning of these sites is critical for the catalytic reaction to occur. The substrate (RNA in this case) is drawn into the cleft by the active sites. By doing so, it puts the substrate molecule under stress, causing the reaction to proceed more readily.

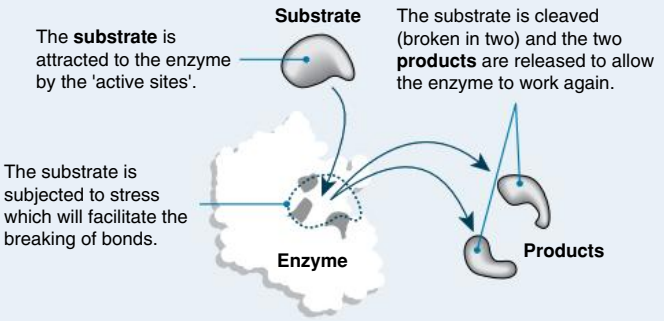


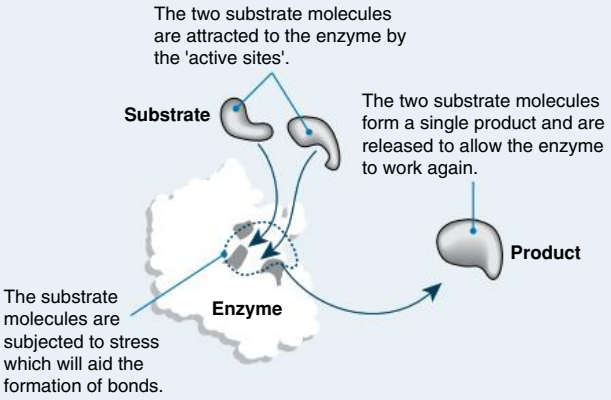
Substrate molecule: Substrate molecules are the chemicals that an enzyme acts on. They are drawn into the cleft of the enzyme.

Active site: These attraction points draw the substrate to the enzyme's surface. Substrate molecule(s) are positioned in a way to promote a reaction: either joining two molecules together or splitting up a larger one (as in this case).

Enzyme molecule: The complexity of the active site is what makes each enzyme so specific for the substrate it acts on.

Source: After *Biochemistry*, (1981) by Lubert Stryer





Catabolic reactions

Some enzymes can cause a single substrate molecule to be drawn into the active site. Chemical bonds are broken, causing the substrate molecule to break apart to become two separate molecules. Catabolic reactions break down complex molecules into simpler ones and involve a net release of energy, so they are called exergonic. **Examples**: *hydrolysis*, *cellular respiration*.

Anabolic reactions

Some enzymes can cause two substrate molecules to be drawn into the active site. Chemical bonds are formed, causing the two substrate molecules to form bonds and become a single molecule. Anabolic reactions involve a net use of energy (they are endergonic) and build more complex molecules and structures from simpler ones. **Examples:** *protein synthesis*, *photosynthesis*.

1. Explain what is meant by the active site of an enzyme and relate it to the enzyme's tertiary structure:

2. What might happen to an enzyme's activity if a mutation caused a change to the shape of the active site?

3. Distinguish between **catabolism** and **anabolism**, giving an example of each:

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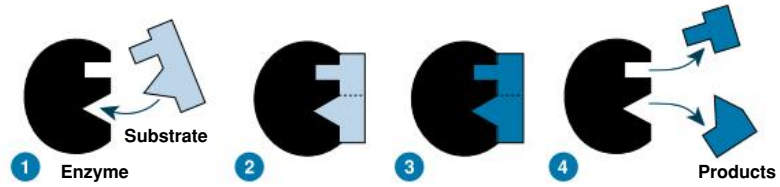
19 How Enzymes Work

Key Idea: Enzymes increase the rate of biological reactions by lowering the reaction's activation energy.

Chemical reactions in cells are accompanied by energy changes. The amount of energy released or taken up is directly related to the tendency of a reaction to run to completion (for all the reactants to form products). Any reaction needs to raise the energy of the substrate to an unstable transition state before the reaction will proceed (below left). The amount of energy needed to do this is the **activation energy** (*Ea*). Enzymes lower the *Ea* by destabilising bonds in the substrate so that it is more reactive. The current 'induced-fit' model of enzyme function is supported by studies of enzyme inhibitors, which show that enzymes are flexible and change shape when interacting with the substrate.

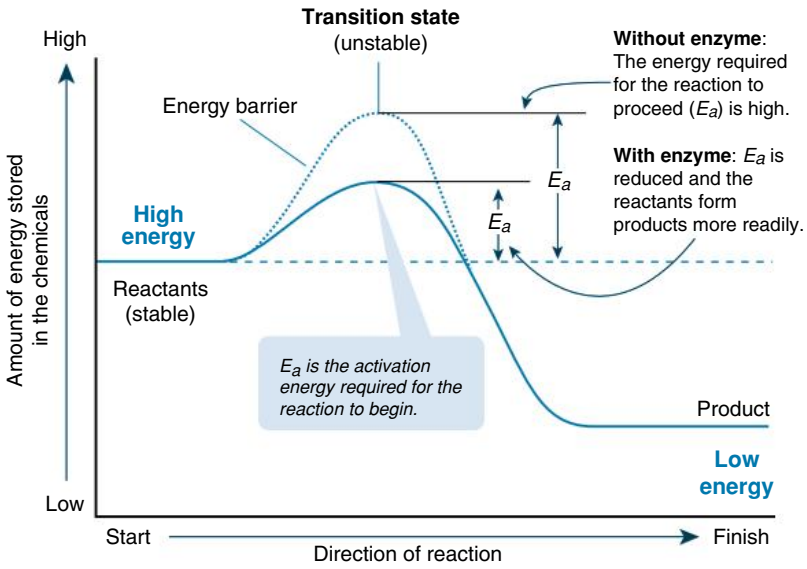
How Enzymes Work

The **lock and key** model proposed earlier last century suggested that the (perfectly fitting) substrate was simply drawn into a matching cleft on the enzyme molecule (below). This model was supported by early X-ray crystallography but has since been modified to recognise the flexibility of enzymes (the **induced fit** model, right).



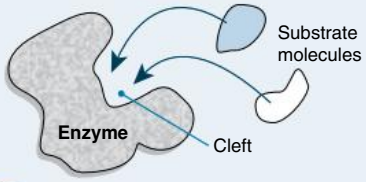
Lowering the Activation Energy

The presence of an enzyme simply makes it easier for a reaction to take place. All catalysts speed up reactions by influencing the stability of bonds in the reactants. They may also provide an alternative reaction pathway, thus lowering the activation energy (E_a) needed for a reaction to take place (see the graph below).

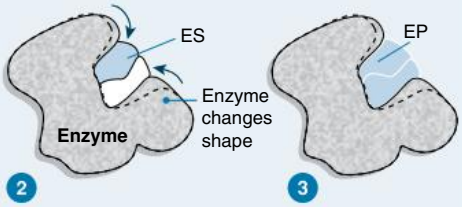


The Current Model: Induced Fit

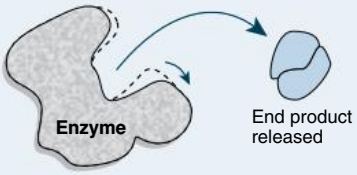
An enzyme's interaction with its substrate is best regarded as an induced fit (below). The shape of the enzyme changes when the substrate fits into the cleft. The reactants become bound to the enzyme by weak chemical bonds. This binding can weaken bonds within the reactants themselves, allowing the reaction to proceed more readily.



1 Two substrate molecules are drawn into the cleft of the enzyme.



The enzyme changes shape as the substrate molecules bind in an enzyme-substrate complex (ES). The enzyme-substrate interaction results in an intermediate enzyme product (EP) complex.



4 The end product is released and the enzyme returns to its previous shape.

1. Explain how enzymes act as **biological catalysts**:

2. Describe the key features of the '**lock and key**' model of enzyme action and explain its deficiencies as a working model:

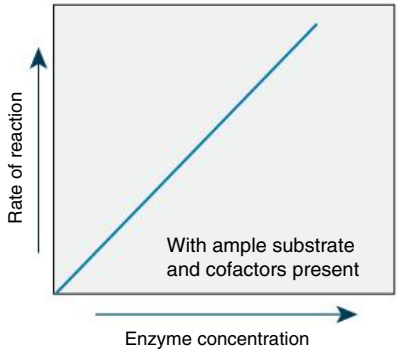
3. Describe the current '**induced fit**' model of enzyme action, explaining how it differs from the lock and key model:

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20 Enzyme Reaction Rates

Key Idea: Enzymes operate most effectively within a narrow range of conditions. The rate of enzyme catalysed reactions is influenced by both enzyme and substrate concentration.

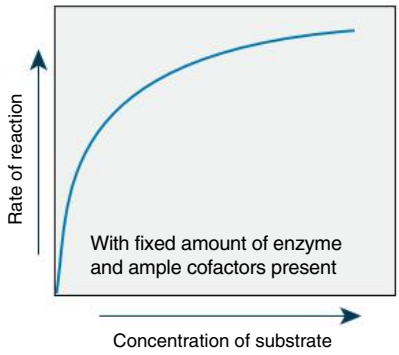
Enzymes usually have an optimum set of conditions (e.g. of pH and temperature) under which their activity is greatest. Many plant and animal enzymes show little activity at low temperatures. Enzyme activity increases with increasing temperature, but falls off after the optimum temperature is exceeded and the enzyme is denatured (loses its tertiary structure and biological activity). Extremes in pH can also cause denaturation. Within their normal operating conditions, enzyme reaction rates are influenced by enzyme and substrate concentration in a predictable way (below). In the four graphs below, the *rate of reaction* or *degree of enzyme activity* is plotted against each of four factors that affect enzyme function.



1. Enzyme concentration

(a) Describe the change in the rate of reaction when the enzyme concentration is increased (assuming there is plenty of the substrate present):

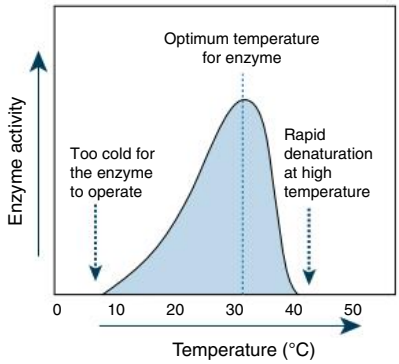
(b) Suggest how a cell may vary the amount of enzyme present in a cell:



2. Substrate concentration

(a) Describe the change in the rate of reaction when the substrate concentration is **increased** (assuming a fixed amount of enzyme and ample cofactors):

(b) Explain why the rate changes the way it does:

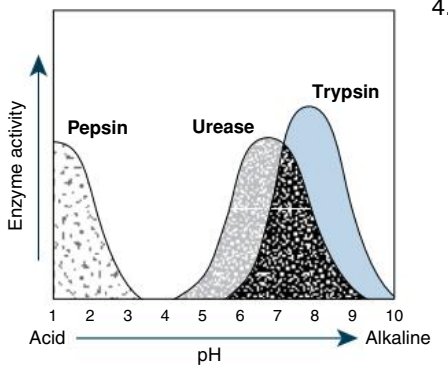


3. Temperature

Higher temperatures speed up all reactions, but few enzymes can tolerate temperatures higher than 50–60°C. The rate at which enzymes are **denatured** (change their shape and become inactive) increases with higher temperatures.

(a) Describe what is meant by an *optimum temperature* for enzyme activity:

(b) Explain why most enzymes perform poorly at low temperatures:



4. Acidity and alkalinity (pH)

Like all proteins, enzymes are **denatured** by *extremes* of **pH** (very acid or alkaline). Within these extremes, most enzymes are still influenced by pH. Each enzyme has a preferred pH range for optimum activity.

(a) State the optimum pH for each of the enzymes:

Pepsin: _____ Trypsin: _____ Urease: _____

(b) Pepsin acts on proteins in the stomach. Explain how its optimum pH is suited to its working environment:

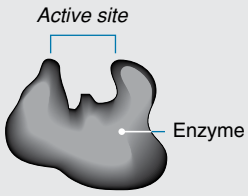
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21 Enzyme Cofactors and Inhibitors

Key Idea: Some enzymes can act as catalysts on their own, but others need the addition of another molecule to function. Enzyme activity can also be reduced by inhibitors.

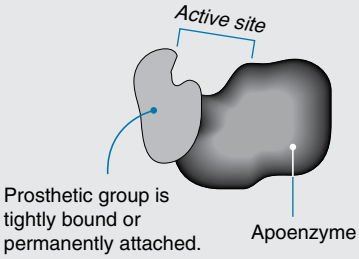
Some enzymes (e.g. pepsin) consist of only protein. Other enzymes require the addition of extra non-protein components to complete their catalytic properties. In these cases, the protein portion is called the **apoenzyme** and the additional chemical component is called a **cofactor**. Neither one has catalytic activity on its own. Cofactors may be organic molecules (e.g. vitamin C) or inorganic ions (e.g. Ca^{2+}) and may be tightly or loosely bound to the enzyme. Enzymes may be deactivated, temporarily or permanently, by chemicals called inhibitors, which may compete with the substrate for the active site or render the active site non-functional.

Protein-only enzymes



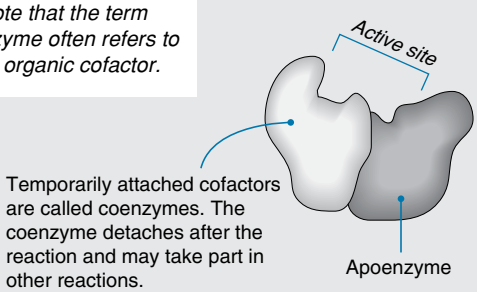
No cofactor
Functional enzyme consists of only protein
e.g. lysozyme, pepsin

Conjugated protein enzymes



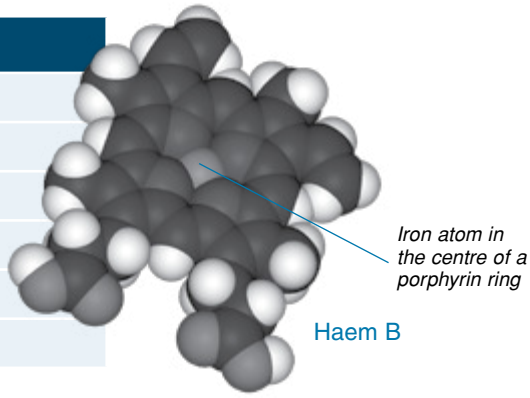
Prosthetic group required
Contains apoenzyme (protein) plus a prosthetic group
e.g. flavoprotein + FAD

Note that the term coenzyme often refers to any organic cofactor.



Coenzyme required
Contains apoenzyme (protein) plus a coenzyme (non-protein)
e.g. dehydrogenases + NAD

Enzyme cofactor	Enzyme
Cupric (copper ion)	Cytochrome oxidase
Ferrous or ferric (iron ion)	Catalase and cytochrome (via heme)
Selenium	Glutathione peroxidase
Magnesium	Glucose 6-phosphatase
Flavin	NADH dehydrogenase
Haem L (derived from haem B)	Peroxidases, e.g. thyroid peroxidase



1. Describe the general role of **cofactors** in enzyme activity: _____

2. Explain exactly how cofactors enable an enzyme's catalytic activity: _____

3. Distinguish between the apoenzyme and the cofactor: _____

4. Identify the two broad categories of cofactors and describe an example of each: _____

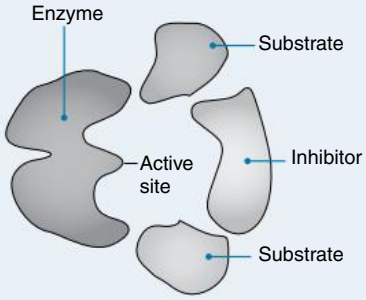
5. With reference to enzyme activity, describe the importance of adequate vitamin and mineral intake in the diet:

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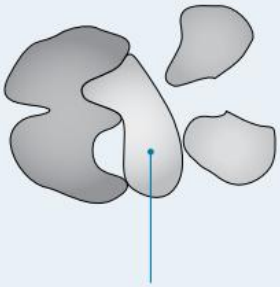
Competitive Inhibition

Competitive inhibitors compete with the normal substrate for the enzyme's active site.

Competitive inhibitors compete directly with the substrate for the active site, and their effect can be overcome by increasing the substrate concentration.

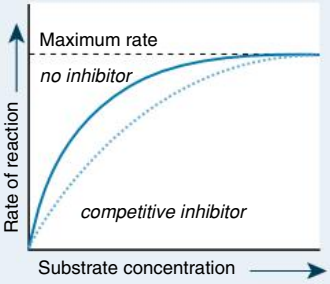


1 Inhibitor is present in the cell (or solution) with the substrate



2 Inhibitor temporarily binds to the active site, blocking it so that the substrate cannot bind

Fig.1 Effect of competitive inhibition on enzyme reaction rate at different substrate concentration

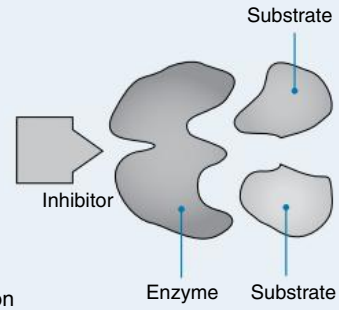


Non-competitive Inhibition

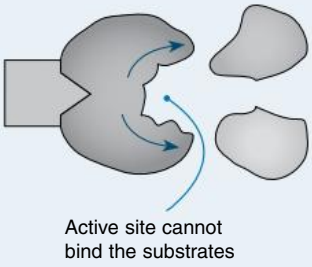
Non-competitive inhibitors bind with the enzyme at a site other than the active site.

They inactivate the enzyme by altering its shape so that the substrate and enzyme can no longer interact.

Non-competitive inhibition cannot be overcome by increasing the substrate concentration.

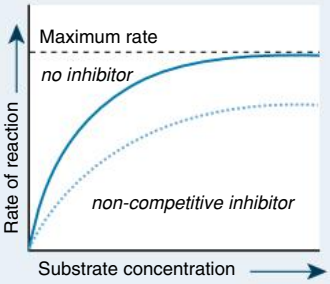


1 Without the inhibitor bound, the enzyme can bind the substrate



2 When the inhibitor binds, the enzyme changes shape.

Fig.2 Effect of non-competitive inhibition on enzyme reaction rate at different substrate concentration



6. Distinguish between **competitive** and **non-competitive** inhibition: _____

7. (a) Compare and contrast the effect of competitive and non-competitive inhibition on the relationship between the substrate concentration and the rate of an enzyme controlled reaction (figs 1 and 2 above):

(b) How could you distinguish between competitive and non-competitive inhibition in an isolated system?

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22 Achieving Metabolic Efficiency

Key Idea: Metabolic pathways are linked biochemical reactions that occur within organisms to maintain life.

Each step in a metabolic pathway relies on the completion of a previous step and each step is controlled by specific **enzymes**. The end product of one enzyme-controlled step provides the substrate for the next step, so failure of one step causes failure of all later steps. Metabolic pathways are tightly controlled to prevent energy being wasted. This energy conservation is termed **metabolic efficiency**. Metabolic reactions are often localised within regions of a cell, or within specific organelles so that all the necessary components of a metabolic pathway are kept together.

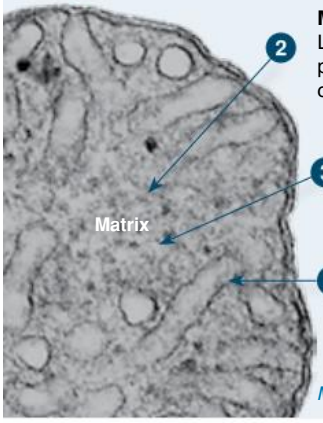
Achieving Efficiency by Compartmentalisation

To increase metabolic efficiency, regions within a cell or an organelle are **compartmentalised** (separated) by membranes. Particular metabolic reactions are restricted to certain regions where all the necessary metabolic components are located. Compartmentalisation prevents interference between different reaction pathways and enables radically different reaction environments to be accommodated within different organelles.

Example: Cellular Respiration in the Mitochondrion

The membrane system of the mitochondrion divides it into several regions. Glycolysis takes place outside of the mitochondrion, in the cell's cytoplasm, but the remaining steps take place in different specialised regions of the mitochondrion. This helps to regulate movement of substrates and end-products and therefore reaction rates, increasing efficiency of the process (below).

1. Cytoplasm (outside the mitochondrion): Glycolysis



2. **Matrix**: Link reaction. Link reaction enzymes (e.g. pyruvate dehydrogenase complex) are in the matrix.
3. **Matrix**: Krebs cycle. Krebs cycle enzymes (e.g. fumarase) are in the matrix.
4. **Cristae**: Electron transport chain. Membrane-bound enzymes include ATP synthase

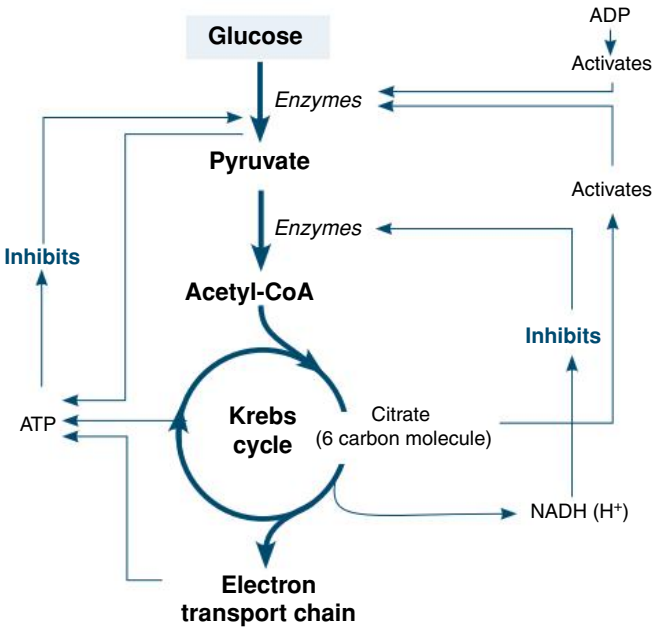
Mitochondrion

WMU

Achieving Efficiency by Inhibition

Many metabolic pathways are controlled by **feedback inhibition** (negative feedback loop). The pathway is stopped when a build-up of end product (or certain intermediate products) occurs. The build-up stops the enzymes in the pathway from working and allows cells to shut down a pathway when it is not needed. This conserves the cell's energy, so it is not manufacturing products it does not need.

Both linear pathways (e.g. glycolysis), and cyclic pathways (e.g. the Krebs cycle) and can be regulated this way (below).



1. What does metabolic efficiency mean? _____

2. Describe how cells achieve metabolic efficiency through:

   (a) **Compartmentailisation**: _____

   (b) **Feedback inhibition**: _____

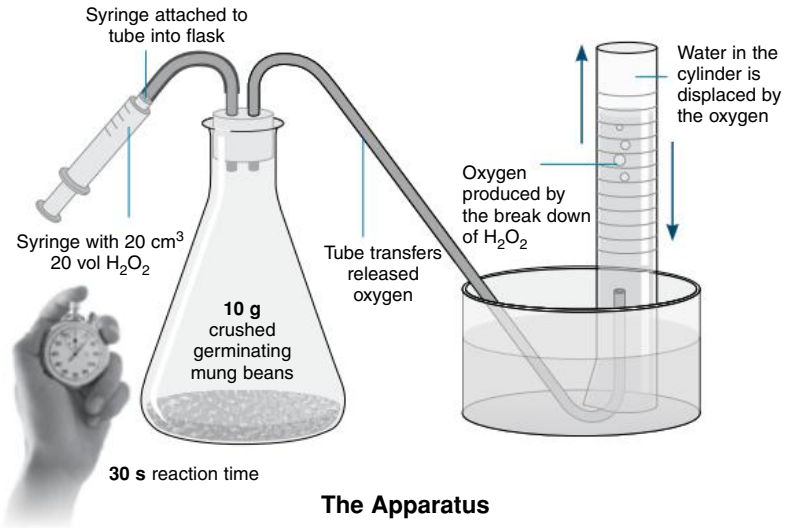
3. What would happen if cells could not regulate their metabolic pathways? _____

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23 Catalase Activity in Germinating Seeds

Key Idea: Catalase activity can be measured in germinating seeds. Activity changes with stage of germination.

Enzyme activity can be measured easily in simple experiments. This activity describes an experiment in which germinating seeds of different ages were tested for their level of **catalase activity** using hydrogen peroxide solution as the substrate and a simple apparatus to measure oxygen production (see background). Completing this activity, which involves a critical evaluation of the second-hand data provided, will help to prepare you for making your own similar investigations.



The Apparatus

In this experiment, 10 g germinating mung bean seeds (0.5, 2, 4, 6, or 10 days old) were ground by hand with a mortar and pestle and placed in a conical flask as above. There were six trials at each of the five seedling ages. With each trial, 20 cm^3 of 20 vol H_2O_2 was added to the flask at time 0 and the reaction was allowed to run for 30 seconds. The oxygen released by the decomposition of the H_2O_2 by catalase in the seedlings was collected via a tube into an inverted measuring cylinder. The volume of oxygen produced is measured by the amount of water displaced from the cylinder. The results from all trials are tabulated below:

The Aim

To investigate the effect of germination age on the level of catalase activity in mung beans.

Background

Germinating seeds are metabolically very active and this metabolism inevitably produces reactive oxygen species, including hydrogen peroxide (H_2O_2). H_2O_2 helps germination by breaking dormancy, but it is also toxic. To counter the toxic effects of H_2O_2 and prevent cellular damage, germinating seeds also produce **catalase**, an enzyme that catalyses the breakdown of H_2O_2 to water and oxygen.

A class was divided into six groups with each group testing the seedlings of each age. Each group's set of results (for 0.5, 2, 4, 6, and 10 days) therefore represents one trial.

Stage of germination (days) / Trial #	Volume of oxygen collected after 30 s (cm^3)								Mean rate ($cm^3\ s^{-1}\ g^{-1}$)
	1	**2**	**3**	**4**	**5**	**6**	Mean	Standard deviation	
0.5	9.5	10	10.7	9.5	10.2	10.5			
2	36.2	30	31.5	37.5	34	40			
4	59	66	69	60.5	66.5	72			
6	39	31.5	32.5	41	40.3	36			
10	20	18.6	24.3	23.2	23.5	25.5			

1. Write the equation for the catalase reaction with hydrogen peroxide: _____

2. Complete the table above to summarise the data from the six trials:

   (a) Calculate the mean volume of oxygen for each stage of germination and enter the values in the table.

   (b) Calculate the standard deviation for each mean and enter the values in the table (you may use a spreadsheet).

   (c) Calculate the mean rate of oxygen production in cm^3 per second per gram. For the purposes of this exercise, assume that the weight of germinating seed in every case was 10.0 g.

3. In another scenario, group (trial) #2 obtained the following measurements for volume of oxygen produced: 0.5 d: 4.8 cm^3, 2 d: 29.0 cm^3, 4 d: 70 cm^3, 6 d: 30.0 cm^3, 10 d: 8.8 cm^3 (pencil these values in beside the other group 2 data set).

   (a) Describe how group 2's new data compares with the measurements obtained from the other groups: _____

   _____

   (b) Describe how you would approach a reanalysis of the data set incorporating group 2's new data: _____

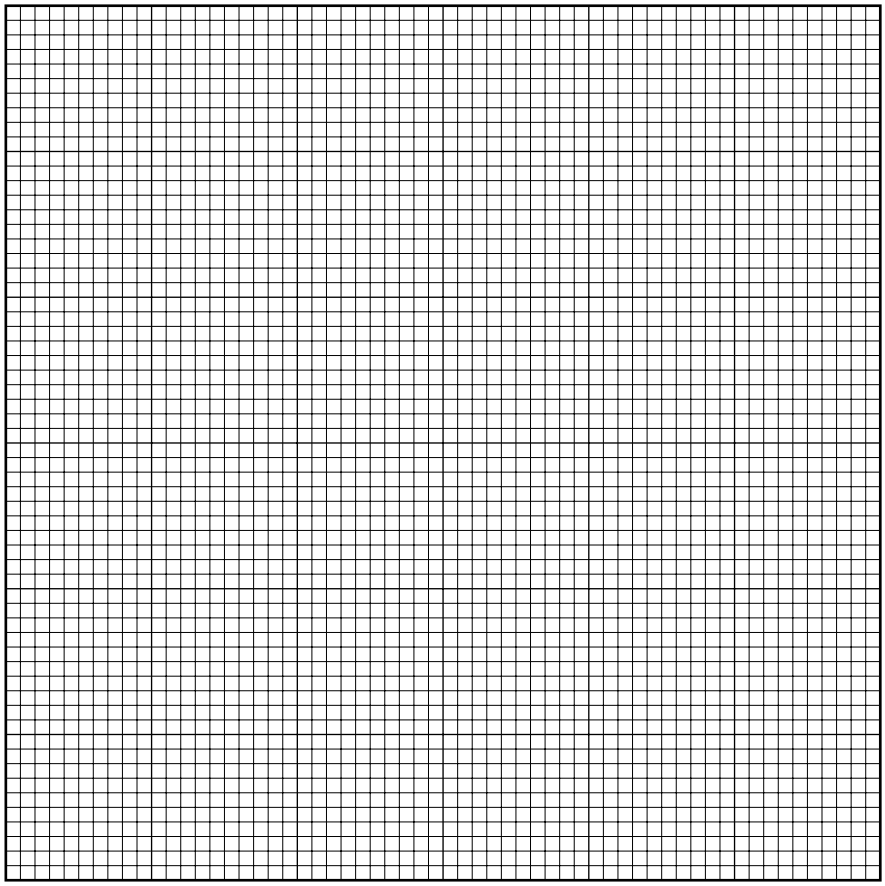
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   _____

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(c) Explain the rationale for your approach

4. Use the tabulated data to plot an appropriate graph of the results on the grid provided:



5. (a) Describe the trend in the data:

(b) Explain the relationship between stage of germination and catalase activity shown in the data:

6. Describe any potential sources of errors in the apparatus or the procedure:

7. Describe two things that might affect the validity of findings in this experimental design:

8. Describe one improvement you could make to the experiment in order to generate more reliable data:

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24 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its correct definition, as identified by its preceding letter code.

activation energy .....

active site .....

amino acids .....

anabolic reactions .....

carbohydrates .....

catabolic reactions .....

catalyst .....

cofactor .....

condensation .....

denaturation .....

endergonic .....

enzymes .....

exergonic .....

fibrous proteins .....

hydrolysis .....

globular proteins .....

hydrogen bond .....

induced fit model .....

lipids .....

monosaccharides .....

nucleic acid .....

optimum (for enzyme) .....

polysaccharide .....

primary structure .....

protein .....

proteome .....

tertiary structure .....

A Substance required by an enzyme to enable its catalytic function.

B Proteins with a rod or wire-like structure that are important in structure or storage.

C Water soluble proteins with a spherical tertiary structure. They are involved in many cellular functions including as catalysts and in transport and regulation.

D Reactions that break up larger molecules into their component parts.

E The structure of a protein maintained by disulfide bonds and hydrophilic and hydrophobic interactions.

F A class of organic compounds with an oily, greasy, or waxy consistency. Important as energy storage molecules and as components of cellular membranes.

G An information-carrying macromolecule composed of chains of nucleotides.

H These are biological catalysts.

I Organic molecules consisting only of carbon, hydrogen and oxygen that serve as structural components in cells and as energy sources.

J Any reagent that increases the rate of a chemical reaction but is itself not consumed by the reaction.

K Organic compounds, usually linear polymers, made of amino acids linked together by peptide bonds.

L The region of an enzyme responsible for substrate binding and reaction catalysis.

M Reactions that release energy are called this.

N Reactions that build larger molecules from smaller ones are called this.

O The energy that must be overcome in order for a chemical reaction to occur.

P The loss of a protein's three dimensional functional structure is called this.

Q A general term for a reaction in which water is released.

R The splitting of a molecule into smaller components by addition of a water molecule.

S Carbohydrate monomers. Examples include fructose and glucose.

T The sequence of amino acids in a polypeptide.

U The sum total of all proteins in an organism.

V The currently accepted model for enzyme function.

W Reactions that require energy to proceed are called this.

X The conditions which an enzyme works best at (e.g. a specific pH or temperature).

Y The building blocks of proteins.

Z Weak attractive interaction between polar molecules.

AA Long carbohydrate molecules made up of monosaccharide units joined together by glycosidic bonds.

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QLD As required
VCE Unit 3 (1)
WA ATAR Unit 2
SA Theme O

Cellular Energetics

Key terms

absorption spectrum
accessory pigment
acetyl coA
action spectrum
anaerobic metabolism
ATP
ATPase
Calvin cycle
cellular respiration
chemiosmosis
chloroplast
chlorophyll
cristae
electron transport chain
ethanol
fermentation
glycolysis
grana
Krebs cycle
lactic acid
light dependent phase
matrix
mitochondrion
NAD/NADP
oxidative phosphorylation
photolysis
photosynthesis
ribulose bisphosphate
RuBisCo
stroma
substrate level phosphorylation
thylakoid discs

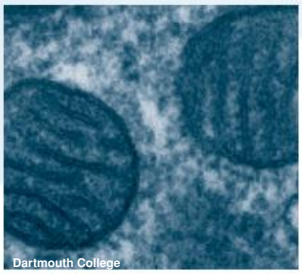


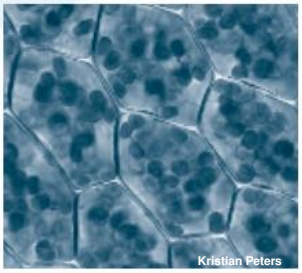
Role of ATP

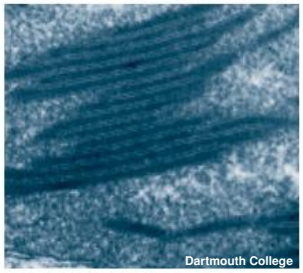
Learning aims and skills

Activity number

- ☐ 1 Explain the universal role of ATP in metabolism, as illustrated by examples, e.g. active transport, anabolic reactions, movement, and thermoregulation. — 25
- ☐ 2 Describe the structure of ATP. Describe its synthesis from ADP and inorganic phosphate (Pi) and explain how it stores and releases its energy. — 26
- ☐ 3 Compare and contrast cellular respiration and photosynthesis as energy transformation processes. — 26







Cellular respiration

Learning aims and skills

Activity number

- ☐ 4 Describe the structure of a mitochondrion, identifying the matrix and cristae. Identify the location of the main steps in the complete oxidation of glucose: glycolysis, the link reaction, Krebs cycle, and electron transport chain. — 27
- ☐ 5 Describe glycolysis and recognise it as the major anaerobic pathway in cells (the starting point for both respiratory and fermentation pathways). State the net yield of ATP and $NADH_2$ from glycolysis. — 28
- ☐ 6 Describe the complete oxidation of glucose to CO_2, including reference to: — 28 29
  - The conversion of pyruvate to acetyl-coenzyme A.
  - The stepwise oxidation of intermediates in the Krebs cycle.
  - Generation of ATP by chemiosmosis in the electron transport chain.
  - The role of oxygen as the terminal electron acceptor.
- ☐ 7 Describe fermentation in mammalian muscle and in yeast, identifying the final electron acceptor in each case. Compare and explain the differences in the yields of ATP from aerobic respiration and from fermentation. — 30 31

Photosynthesis

Learning aims and skills

Activity number

- ☐ 8 Describe the structure and role of chloroplasts. Explain the role of chlorophyll *a* and *b*, and accessory pigments (e.g. carotenoids) in light capture by green plants. — 32 33
- ☐ 9 Explain what is meant by the terms absorption spectrum and action spectrum with respect to light absorbing pigments.
- ☐ 10 Describe and explain the light dependent phase of photosynthesis, including reference to the location and role of the photosystems, the generation of ATP and NADPH, and the role of ATPase. — 35
- ☐ 11 Describe and explain the Calvin cycle and the fixation of carbon dioxide using ATP and NADPH in the light independent phase of photosynthesis. Include reference to the reduction of G3P and the regeneration of ribulose bisphosphate. — 36
- ☐ 12 Describe and explain factors affecting photosynthetic rate and yield. — 37

25 The Role of ATP in Cells

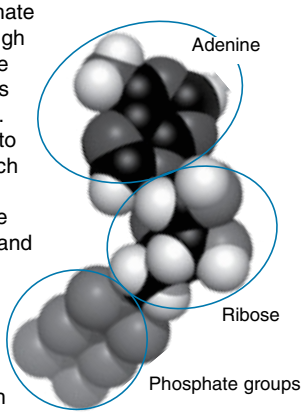
Key Idea: ATP transports chemical energy within the cell for use in metabolic processes.

All organisms require energy to be able to perform the metabolic processes required for them to function and reproduce. This energy is obtained by cellular respiration, a set of metabolic reactions which ultimately convert biochemical energy from 'food' into the nucleotide **adenosine triphosphate** (ATP). ATP is considered to be a universal energy carrier, transferring chemical energy within the cell for use in metabolic processes such as biosynthesis, cell division, cell signalling, thermoregulation, cell mobility, and active transport of substances across membranes.

Adenosine Triphosphate (ATP)

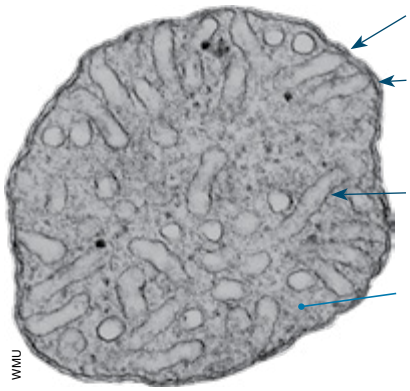
The ATP molecule consists of three components; a purine base (**adenine**), a pentose sugar (**ribose**), and **three phosphate groups** which attach to the 5' carbon of the pentose sugar. The three dimensional structure of ATP is described below.

The bonds between the phosphate groups contain electrons in a high energy state which store a large amount of energy. The energy is released during ATP hydrolysis. Typically, hydrolysis is coupled to another cellular reaction to which the energy is transferred. The end products of the reaction are adenosine diphosphate (ADP) and an inorganic phosphate (Pi). Note that energy is released during the formation of bonds during the hydrolysis reaction, not the breaking of bonds between the phosphates (which requires energy input).



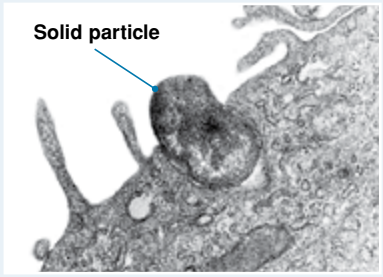
The Mitochondrion

Cellular respiration and ATP production occur in mitochondria. A mitochondrion is bounded by a double membrane. The inner and outer membranes are separated by an intermembrane space, compartmentalising the regions where the different reactions of cellular respiration take place.



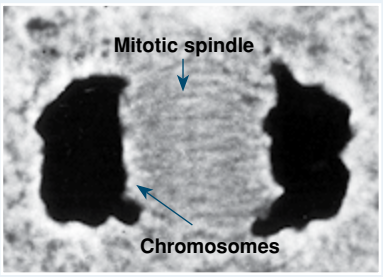
WMU

ATP Powers Metabolism

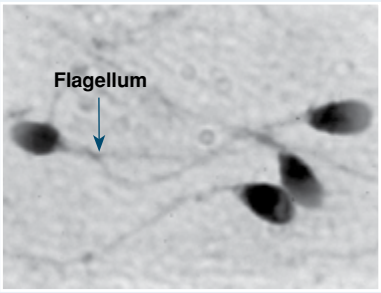


CDC

The energy released from the removal of a phosphate group of ATP is used to actively transport molecules and substances across the cellular membrane. **Phagocytosis** (left), which involves the engulfment of solid particles, is one such example.



Cell division (mitosis), as observed in this onion cell, requires ATP to proceed. Formation of the mitotic spindle and chromosome separation are two aspects of cell division which require energy from ATP hydrolysis to occur.



The hydrolysis of ATP provides the energy for motile cells to achieve movement via a tail-like structure called a flagellum. For example, mammalian sperm must be able to move to the ovum to fertilise it.



EII

Maintaining body temperature requires energy. To maintain body heat, muscular activity increases (e.g. shivering). Cooling requires expenditure of energy too. For example, sweating is an energy requiring process involving secretion from glands in the skin.

1. Explain why organisms need to respire: _____

_____

2. (a) Describe the general role of mitochondria in cell respiration: _____

_____

(b) Explain the importance of compartmentalisation in the mitochondrion: _____

_____

3. Explain why thermoregulation is associated with energy expenditure: _____

_____

_____

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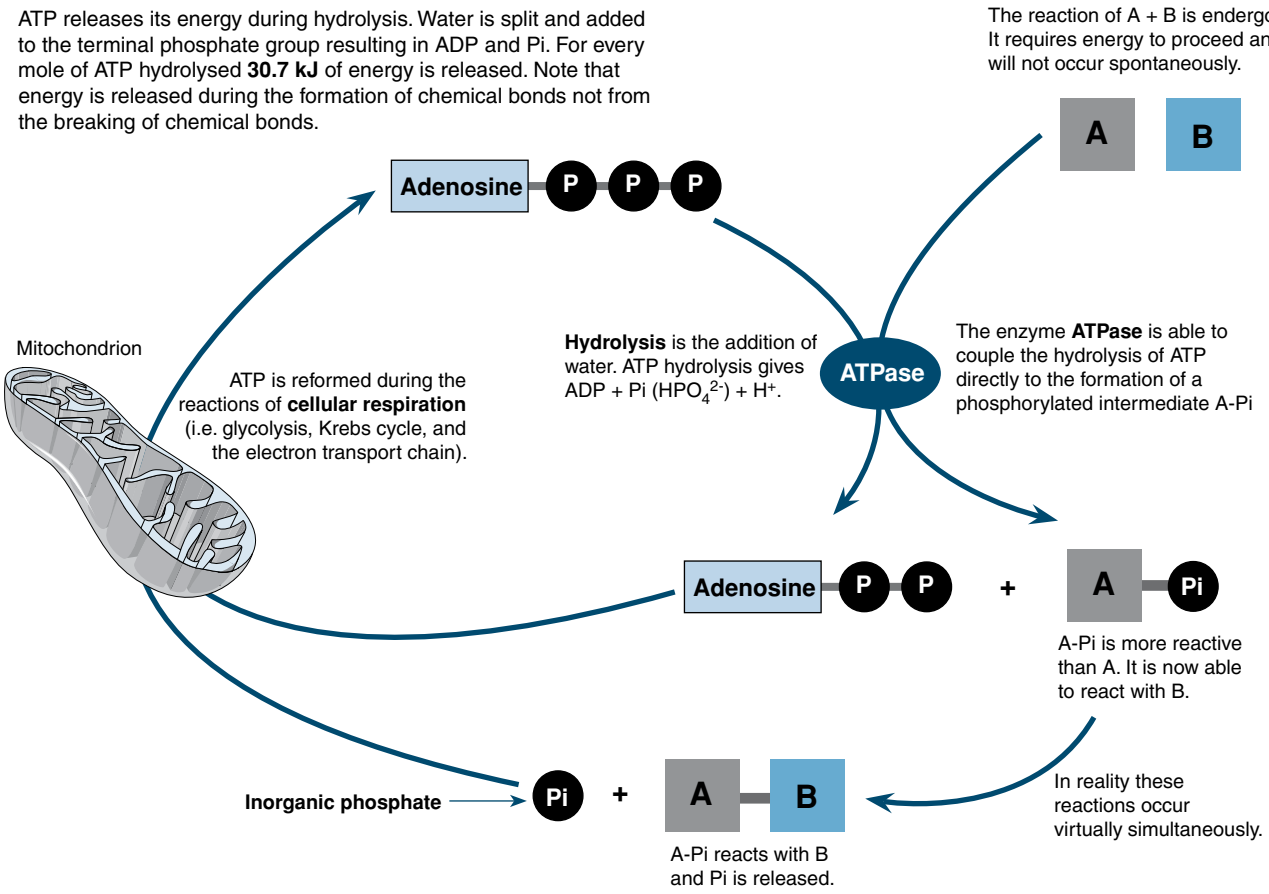
26 ATP and Energy

Key Idea: ATP is the universal energy carrier in cells. Energy is stored in the covalent bonds between phosphate groups.

The molecule ATP (adenosine triphosphate) is the universal energy carrier for the cell. ATP can release its energy quickly by hydrolysis of the terminal phosphate. This reaction is catalysed by the enzyme ATPase. Once ATP has released its energy, it becomes ADP (adenosine diphosphate), a low energy molecule that can be recharged by adding a phosphate. The energy to do this is supplied by the controlled breakdown of respiratory substrates in cellular respiration.

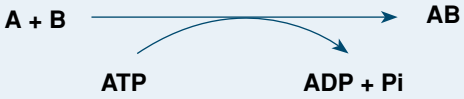
How does ATP Provide Energy?

ATP releases its energy during hydrolysis. Water is split and added to the terminal phosphate group resulting in ADP and Pi. For every mole of ATP hydrolysed **30.7 kJ** of energy is released. Note that energy is released during the formation of chemical bonds not from the breaking of chemical bonds.



Note! The phosphate bonds in ATP are often referred to as high energy bonds. This can be misleading. The bonds contain *electrons in a high energy state* (making the bonds themselves relatively weak). A small amount of energy is required to break the bonds, but when the intermediates recombine and form new chemical bonds a large amount of energy is released. The final product is less reactive than the original reactants.

In many textbooks the reaction series above is simplified and the intermediates are left out:



1. (a) How does ATP supply energy to power metabolism? _____

   _____

   (b) In what way is the ADP/ATP system like a rechargeable battery? _____

   _____

2. What is the immediate source of energy for reforming ATP from ADP? _____
3. Explain the purpose of the folded inner membrane in mitochondria: _____

   _____

   _____

4. Explain why highly active cells (e.g. sperm cells) have large numbers of mitochondria: _____

   _____

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27 ATP Production in Cells

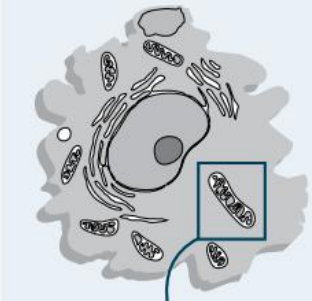
Key Idea: Cellular respiration is the process by which the energy in glucose is transferred to ATP.

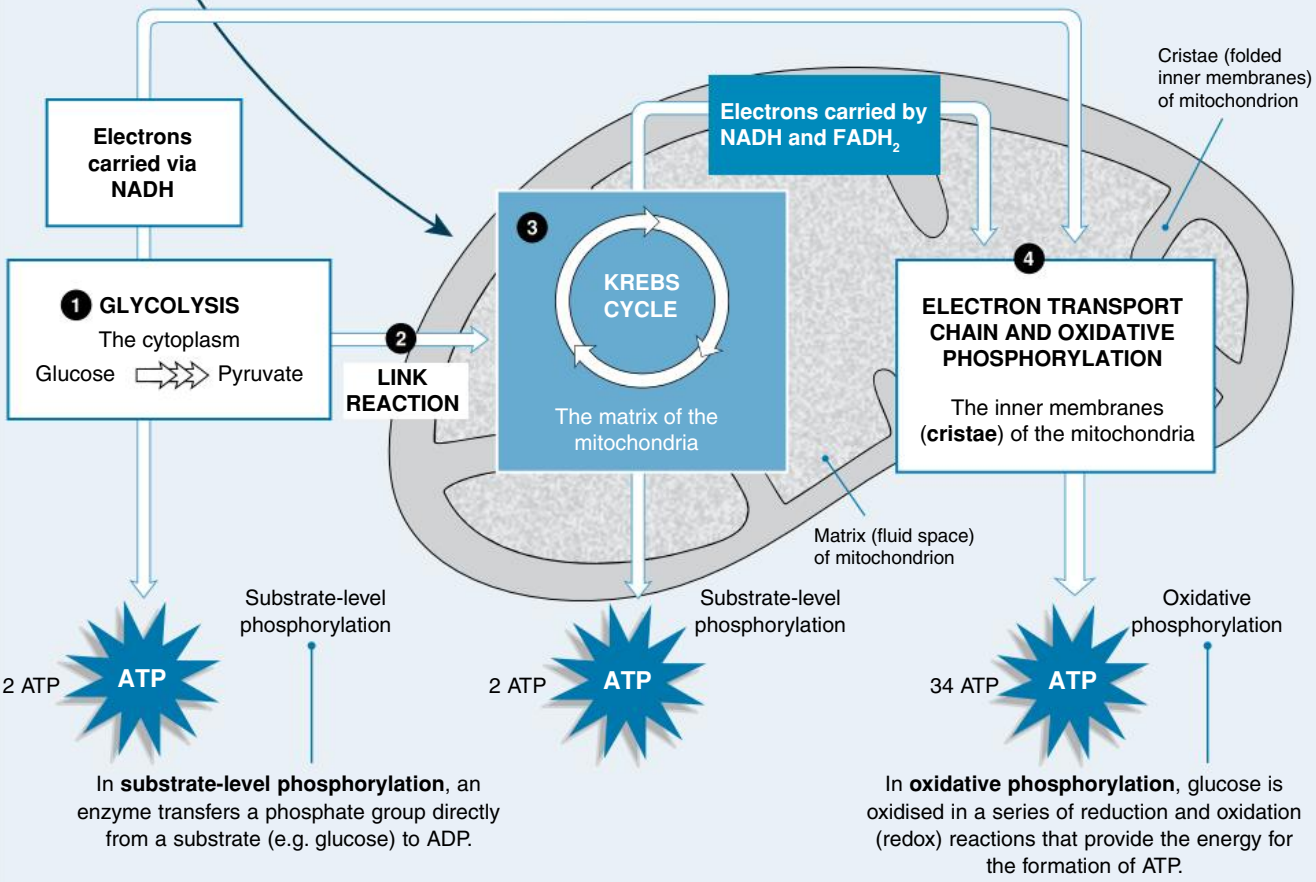
Cellular respiration can be **aerobic** (requires oxygen) or **anaerobic** (does not require oxygen). Some plants and animals can generate ATP anaerobically for short periods of time. Other organisms (anaerobic bacteria) use only anaerobic respiration and live in oxygen-free environments. Cell respiration occurs in the cytoplasm and mitochondria.

An Overview of ATP Production in Cells

Respiration involves three metabolic stages (plus a link reaction) summarised below. The first two stages are the catabolic pathways that decompose glucose and other organic fuels. In the third stage, the electron transport chain accepts electrons from the first two stages and passes these from one electron acceptor to another. The energy released at each stepwise transfer is used to make ATP. The final electron acceptor in this process is molecular oxygen.

1. **Glycolysis**. In the cytoplasm, glucose is broken down into two molecules of pyruvate.
2. **The link reaction**. Pyruvate is split and added to coenzyme A ready to enter the Krebs cycle.
3. **Krebs cycle**. In the mitochondrial matrix, a derivative of pyruvate is decomposed to CO_2.
4. **Electron transport and oxidative phosphorylation**. This occurs in the inner membranes of the mitochondrion and accounts for almost 90% of the ATP generated by respiration.





In **substrate-level phosphorylation**, an enzyme transfers a phosphate group directly from a substrate (e.g. glucose) to ADP.

In **oxidative phosphorylation**, glucose is oxidised in a series of reduction and oxidation (redox) reactions that provide the energy for the formation of ATP.

1. Describe precisely in which part of the cell the following take place:

   (a) Glycolysis: _____

   (b) Krebs cycle reactions: _____

   (c) Electron transport chain: _____

2. How does ATP generation in glycolysis and the Krebs cycle differ from ATP generation via the electron transport chain?

_____

_____

_____

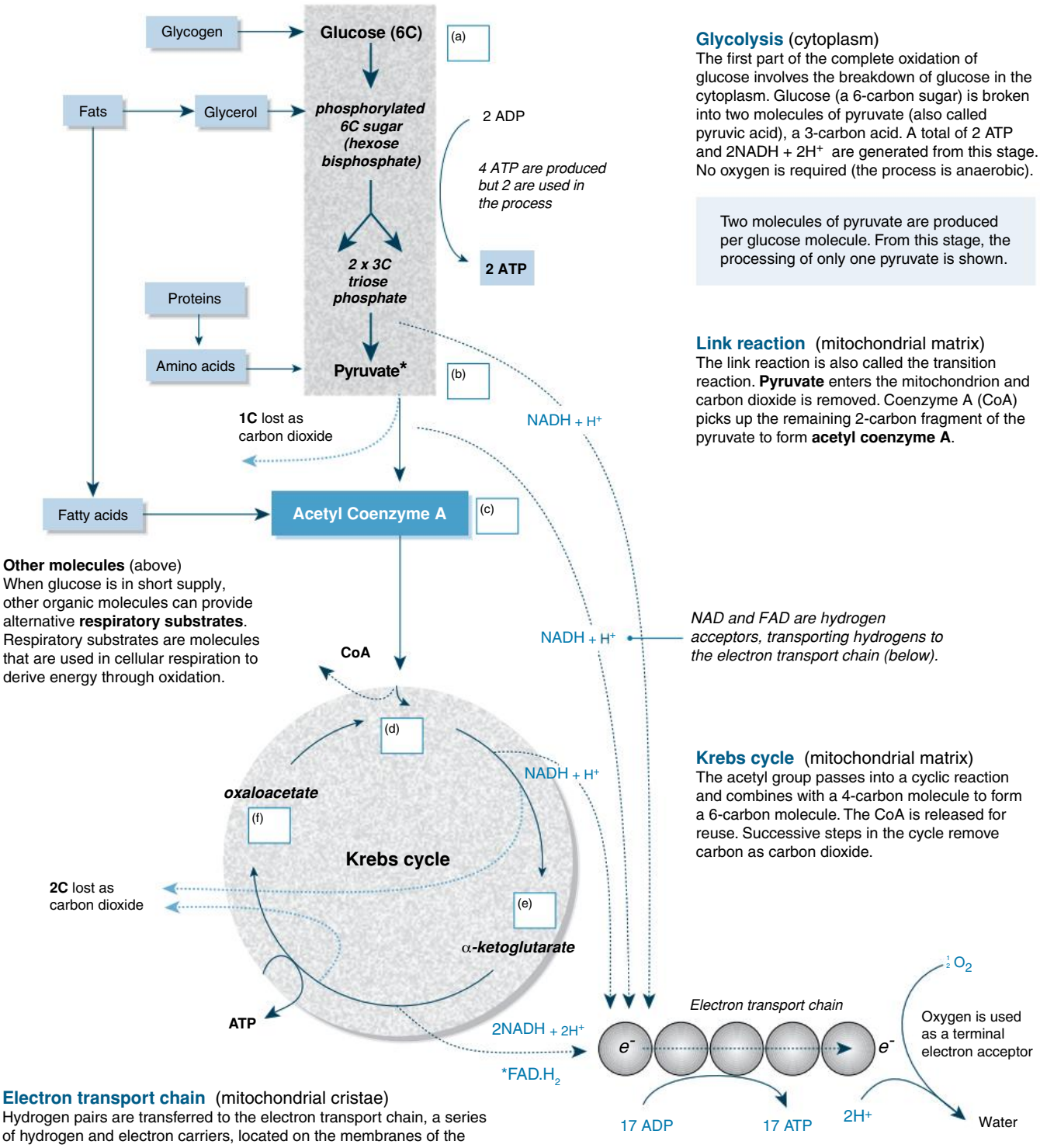
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28 The Biochemistry of Respiration

Key Idea: During cell respiration, the energy in glucose is transferred to ATP in a series of enzyme controlled steps.

The oxidation of glucose is a catabolic, energy yielding pathway. The breakdown of glucose and other organic fuels to simpler molecules is coupled to ATP synthesis. Glycolysis and the Krebs cycle supply electrons to the electron transport chain (ETC), which drives **oxidative phosphorylation**. The conversion of pyruvate (the end product of glycolysis) to **acetyl CoA** links glycolysis to the Krebs cycle. One "turn" of the cycle releases carbon dioxide, forms one ATP, and passes electrons to three NAD^+ and one FAD. Most of the ATP generated in cellular respiration is produced by oxidative phosphorylation when NADH + H^+ and $FADH_2$ donate electrons to the electron carriers in the ETC. At the end of the chain, electrons are passed to molecular oxygen, reducing it to water. Electron transport is coupled to ATP synthesis.



Other molecules (above)
When glucose is in short supply, other organic molecules can provide alternative **respiratory substrates**. Respiratory substrates are molecules that are used in cellular respiration to derive energy through oxidation.

Glycolysis (cytoplasm)
The first part of the complete oxidation of glucose involves the breakdown of glucose in the cytoplasm. Glucose (a 6-carbon sugar) is broken into two molecules of pyruvate (also called pyruvic acid), a 3-carbon acid. A total of 2 ATP and 2NADH + $2H^+$ are generated from this stage. No oxygen is required (the process is anaerobic).

Two molecules of pyruvate are produced per glucose molecule. From this stage, the processing of only one pyruvate is shown.

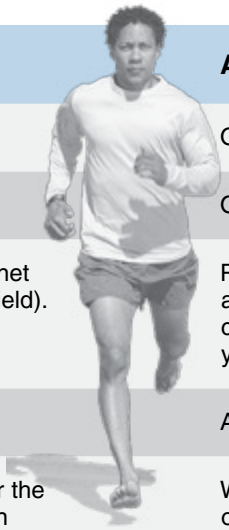
Link reaction (mitochondrial matrix)
The link reaction is also called the transition reaction. **Pyruvate** enters the mitochondrion and carbon dioxide is removed. Coenzyme A (CoA) picks up the remaining 2-carbon fragment of the pyruvate to form **acetyl coenzyme A**.

Krebs cycle (mitochondrial matrix)
The acetyl group passes into a cyclic reaction and combines with a 4-carbon molecule to form a 6-carbon molecule. The CoA is released for reuse. Successive steps in the cycle remove carbon as carbon dioxide.

Electron transport chain (mitochondrial cristae)
Hydrogen pairs are transferred to the electron transport chain, a series of hydrogen and electron carriers, located on the membranes of the cristae. The hydrogens or electrons are passed from one carrier to the next, in a series of redox reactions, losing energy as they go. The energy released in this stepwise process is used to **phosphorylate** ADP to form ATP. Oxygen is the final electron acceptor and is reduced to water (hence the term **oxidative phosphorylation**).
Note FAD enters the electron transport chain at a lower energy level than NAD, and only 2ATP are generated per $FADH_2$.

Total ATP yield per glucose
Glycolysis: 2 ATP, *Krebs cycle*: 2 ATP, *Electron transport*: 34 ATP

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Glycolysis and Fermentation	Aerobic Respiration
Occurs in the cytoplasm of the cell.	Occurs in the mitochondria of eukaryotic cells.
Oxygen is not required.	Oxygen is required.
Glycolysis: glucose is converted to pyruvate with a net production of 2ATP molecules (a very low energy yield).	Pyruvate is converted to carbon dioxide, water, and a further 36 ATP molecules from the Krebs cycle and electron transport chain (a high energy yield per glucose molecule).
Inefficient in terms of energy yield.	An energy efficient process.
In the absence of oxygen, pyruvic acid cannot enter the mitochondrion. It is converted to ethanol and carbon dioxide in plants, and lactic acid in animals.	When oxygen is present, pyruvic acid can be oxidised further via the Krebs cycle and electron transport chain.

1. Summarise the events occurring in each of the following stages of glucose catabolism:

   (a) Glycolysis: _____

   _____

   (b) Link reaction: _____

   _____

   (c) Krebs cycle: _____

   _____

   (d) Electron transport chain: _____

   _____

2. On the diagram on the previous page, state the number of carbon atoms in each of the molecules (a)-(f):

3. Determine how many ATP molecules **per molecule of glucose** are generated during the following stages:

   (a) Glycolysis: _____ (b) Krebs cycle: _____ (c) Electron transport chain: _____ (d) Total: _____

4. Explain what happens to the carbon atoms lost during respiration: _____

   _____

5. Describe the role of each of the following in the complete oxidation of glucose:

   (a) Hydrogen atoms: _____

   _____

   (b) NAD and FAD: _____

   _____

   (c) Oxygen: _____

   _____

   (d) Acetyl coenzyme A: _____

   _____

6. Explain what happens when the supply of glucose for ATP production is limited: _____

   _____

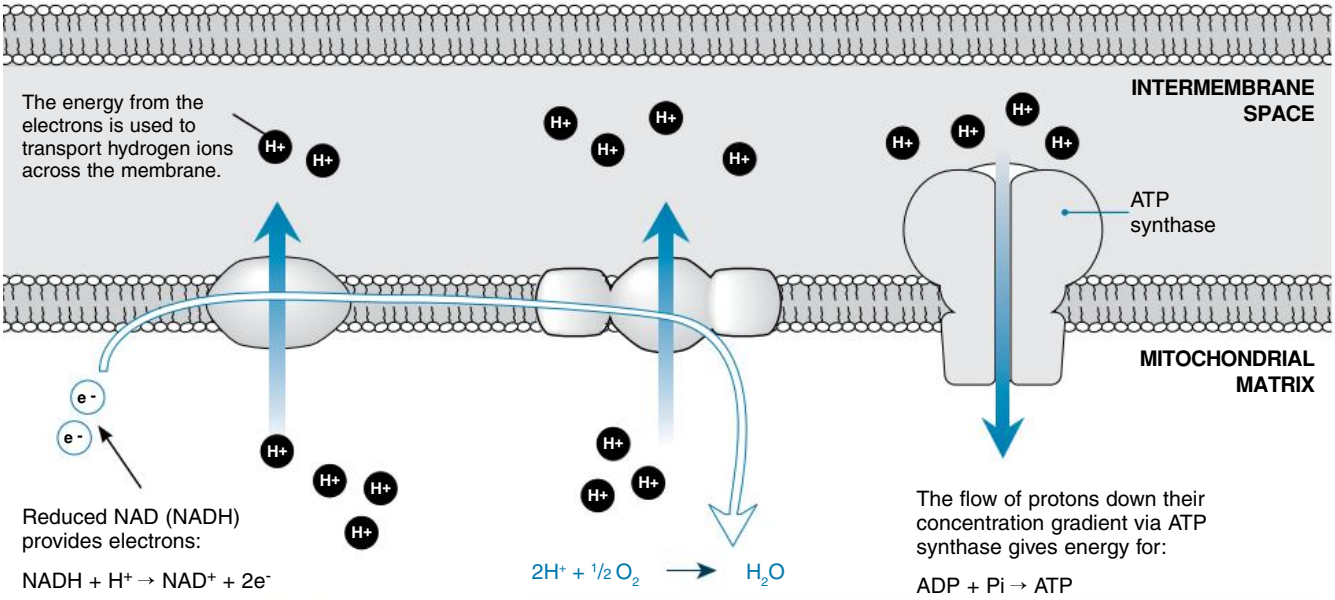
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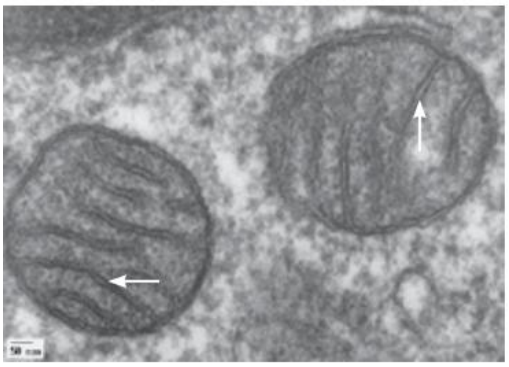
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29 Chemiosmosis

Key Idea: Chemiosmosis is the process in which electron transport is coupled to ATP synthesis.

Chemiosmosis occurs in the membranes of mitochondria, the chloroplasts of plants, and across the plasma membrane of bacteria. Chemiosmosis involves the establishment of a proton (hydrogen) gradient across a membrane. The concentration gradient is used to drive ATP synthesis. Chemiosmosis has two key components: an **electron transport chain** (ETC) sets up a proton gradient as electrons pass along it to a final electron acceptor, and an enzyme called **ATP synthase** uses the proton gradient to catalyse ATP synthesis. In cellular respiration, electron carriers on the inner membrane of the mitochondrion oxidise NADH + H^+ and $FADH_2$. The energy released from this process is used to move protons against their concentration gradient, from the mitochondrial matrix into the space between the two membranes. The return of protons to the matrix via ATP synthase is coupled to ATP synthesis. Similarly, in the chloroplasts of green plants, ATP is produced when protons pass from the thylakoid lumen to the chloroplast stroma via ATP synthase.





Louisa Howard, DEMF

The intermembrane spaces can be seen (arrows) in this transverse section of miotchondria.

The Evidence for Chemiosmosis

The British biochemist Peter Mitchell proposed the chemiosmotic hypothesis in 1961. He proposed that, because living cells have membrane potential, electrochemical gradients could be used to do work, i.e. provide the energy for ATP synthesis. Scientists at the time were skeptical, but the evidence for chemiosmosis was extensive and came from studies of isolated mitochondria and chloroplasts. Evidence included:

- The outer membranes of mitochondria were removed leaving the inner membranes intact. Adding protons to the treated mitochondria increased ATP synthesis.
- When isolated chloroplasts were illuminated, the medium in which they were suspended became alkaline.
- Isolated chloroplasts were kept in the dark and transferred first to a low pH medium (to acidify the thylakoid interior) and then to an alkaline medium (low protons). They then spontaneously synthesised ATP (no light was needed).

1. Summarise the process of chemiosmosis: _____

2. Why did the addition of protons to the treated mitochondria increase ATP synthesis? _____

3. Why did the suspension of isolated chloroplasts become alkaline when illuminated? _____

4. (a) What was the purpose of transferring the chloroplasts first to an acid then to an alkaline medium? _____

   (b) Why did ATP synthesis occur spontaneously in these treated chloroplasts? _____

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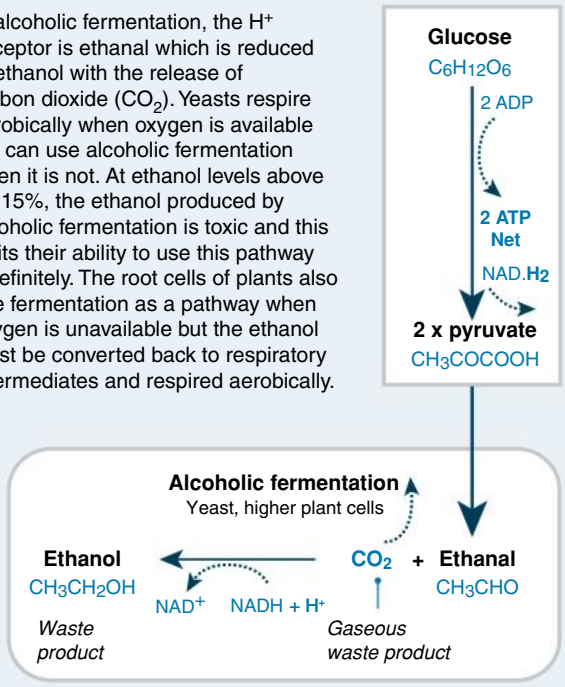
30 Anaerobic Pathways

Key Idea: Glucose can be metabolised aerobically and anaerobically to produce ATP. The ATP yield from aerobic processes is higher than from anaerobic processes.

Aerobic respiration occurs in the presence of oxygen. Organisms can also generate ATP when oxygen is absent by using a molecule other than oxygen as the terminal electron acceptor for the pathway. In alcoholic fermentation in yeasts, the electron acceptor is ethanal. In lactic acid fermentation, which occurs in mammalian muscle even when oxygen is present, the electron acceptor is pyruvate itself.

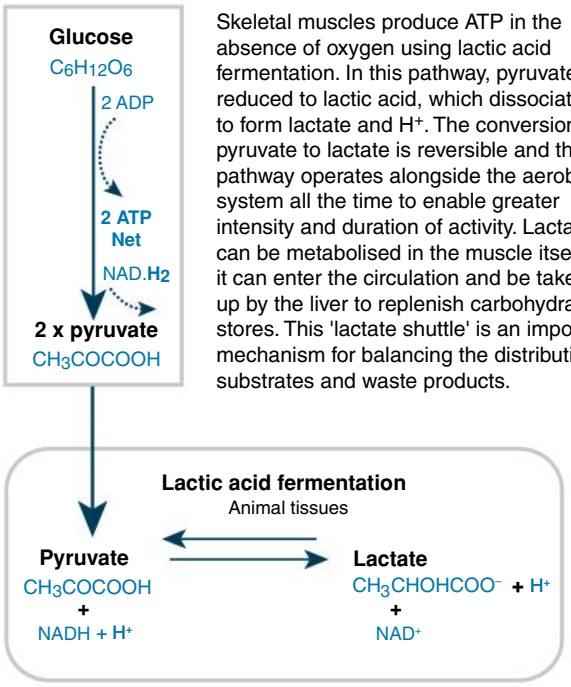
Alcoholic Fermentation

In alcoholic fermentation, the H^+ acceptor is ethanal which is reduced to ethanol with the release of carbon dioxide (CO_2). Yeasts respire aerobically when oxygen is available but can use alcoholic fermentation when it is not. At ethanol levels above 12-15%, the ethanol produced by alcoholic fermentation is toxic and this limits their ability to use this pathway indefinitely. The root cells of plants also use fermentation as a pathway when oxygen is unavailable but the ethanol must be converted back to respiratory intermediates and respired aerobically.



Lactic Acid Fermentation

Skeletal muscles produce ATP in the absence of oxygen using lactic acid fermentation. In this pathway, pyruvate is reduced to lactic acid, which dissociates to form lactate and H^+. The conversion of pyruvate to lactate is reversible and this pathway operates alongside the aerobic system all the time to enable greater intensity and duration of activity. Lactate can be metabolised in the muscle itself or it can enter the circulation and be taken up by the liver to replenish carbohydrate stores. This 'lactate shuttle' is an important mechanism for balancing the distribution of substrates and waste products.





The alcohol and CO_2 produced from alcoholic fermentation form the basis of the brewing and baking industries. In baking, the dough is left to ferment and the yeast metabolises sugars to produce ethanol and CO_2. The CO_2 causes the dough to rise.



Yeasts are used to produce almost all alcoholic beverages (e.g. wine and beers). The yeast used in the process breaks down the sugars into ethanol (alcohol) and CO_2. The alcohol produced is a metabolic by-product of fermentation by the yeast.



Wirtec

The lactate shuttle in vertebrate skeletal muscle works alongside the aerobic system to enable maximal muscle activity. Lactate moves from its site of production to regions within and outside the muscle (e.g. liver) where it can be respired aerobically.

1. Describe the key difference between aerobic respiration and fermentation: _____

_____

2. (a) Refer to page 42 and determine the efficiency of fermentation compared to aerobic respiration: _____ %

   (b) Why is the efficiency of these anaerobic pathways so low? _____

_____

3. Why can't alcoholic fermentation go on indefinitely? _____

_____

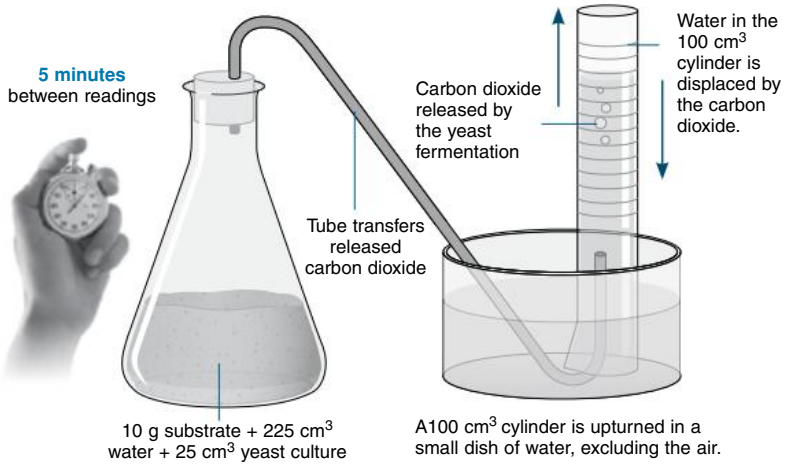
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31 Investigating Yeast Fermentation

Key Idea: Brewer's yeast preferentially uses alcoholic fermentation when there is excess sugar, releasing CO_2, which can be collected as a measure of fermentation rate.

Brewer's yeast is a facultative anaerobe (meaning it can respire aerobically or use fermentation). It will preferentially use alcoholic fermentation when sugars are in excess. One would expect glucose to be the preferred substrate, as it is the starting molecule in cell respiration, but brewer's yeast is capable of utilising a variety of sugars, including disaccharides that can be broken down into single units.



The Apparatus

In this experiment, all substrates tested used the same source culture of 30 g active yeast dissolved in 150 cm^3 of room temperature (24°C) tap water. For each substrate, 25 g of the substrate to be tested was added to 225 cm^3 room temperature (24°C) tap water buffered to pH 4.5. Then 25 cm^3 of source culture was added to the test solution. The control contained yeast solution but no substrate:

Substrate / Time (min)	Group 1: Volume of carbon dioxide collected (cm^3)				
	None	Glucose	Maltose	Sucrose	Lactose
0	0	0	0	0	0
5	0	0	0.8	0	0
10	0	0	0.8	0	0
15	0	0	0.8	0.1	0
20	0	0.5	2.0	0.8	0
25	0	1.2	3.0	1.8	0
30	0	2.8	3.6	3.0	0.5
35	0	4.2	5.4	4.8	0.5
40	0	4.6	5.6	4.8	0.5
45	0	7.4	8.0	7.2	1.0
50	0	10.8	8.9	7.6	1.3
55	0	13.6	9.6	7.7	1.3
60	0	16.1	10.4	9.6	1.3
65	0	22.0	12.1	10.2	1.8
70	0	23.8	14.4	12.0	1.8
75	0	26.7	15.2	12.6	2.0
80	0	32.5	17.3	14.3	2.1
85	0	37.0	18.7	14.9	2.4
90	0	39.9	21.6	17.2	2.6

Substrate / Time (min)	Group 2: Volume of carbon dioxide collected (cm^3)				
	None	Glucose	Maltose	Sucrose	Lactose
90	0	24.4	19.0	17.5	0

The Aim

To investigate the suitability of different mono- and disaccharide sugars as substrates for alcoholic fermentation in yeast.

Background

The rate at which brewer's or baker's yeast (*Saccharomyces cerevisiae*) metabolises carbohydrate substrates is influenced by factors such as temperature, solution pH, and type of carbohydrate available. The literature describes yeast metabolism as optimal in warm, slightly acid environments and this experiment was set up to investigate the pH optimum for the active yeast which is readily available at supermarkets.

Note: High levels of sugars suppress aerobic respiration in yeast, so yeast will preferentially use the fermentation pathway in the presence of excess substrate.

1. Write the equation for the fermentation of glucose by yeast:

   _____

2. Calculate the rate of carbon dioxide production per minute for each substrate in group 1's results:

   (a) None: _____

   (b) Glucose: _____

   (c) Maltose: _____

   (d) Sucrose: _____

   (e) Lactose: _____

3. A second group of students performed the same experiment. Their results are summarised, below left. Calculate the rate of carbon dioxide production per minute for each substrate in group 2's results:

   (a) None: _____

   (b) Glucose: _____

   (c) Maltose: _____

   (d) Sucrose: _____

   (e) Lactose: _____

Experimental design and results adapted from Tom Schuster, Rosalie Van Zyl, & Harold Coller , California State University Northridge 2005

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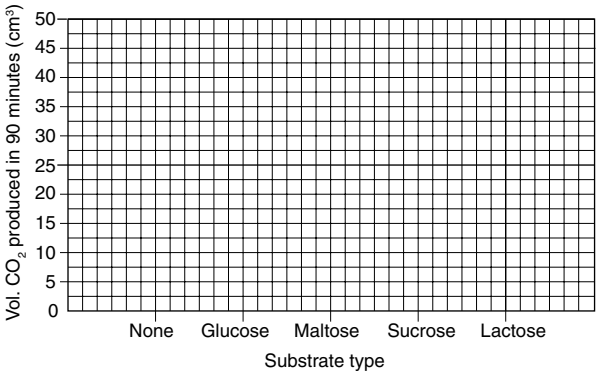
4. What assumptions are being made in this experimental design and do you think they were reasonable?

5. Use the tabulated data to plot an appropriate graph of group 1's results on the grid provided:

6. (a) Summarise the results of group 1's fermentation experiment:

(b) Explain the findings based on your understanding of cellular respiration and carbohydrate chemistry:

7. (a) Plot a column chart to compare the results of the two groups in the volume of CO_2 collected after 90 minutes for each substrate (axes have been completed):



(b) Compare the results of the two groups:

(c) Provide a probable explanation for any differences in the results:

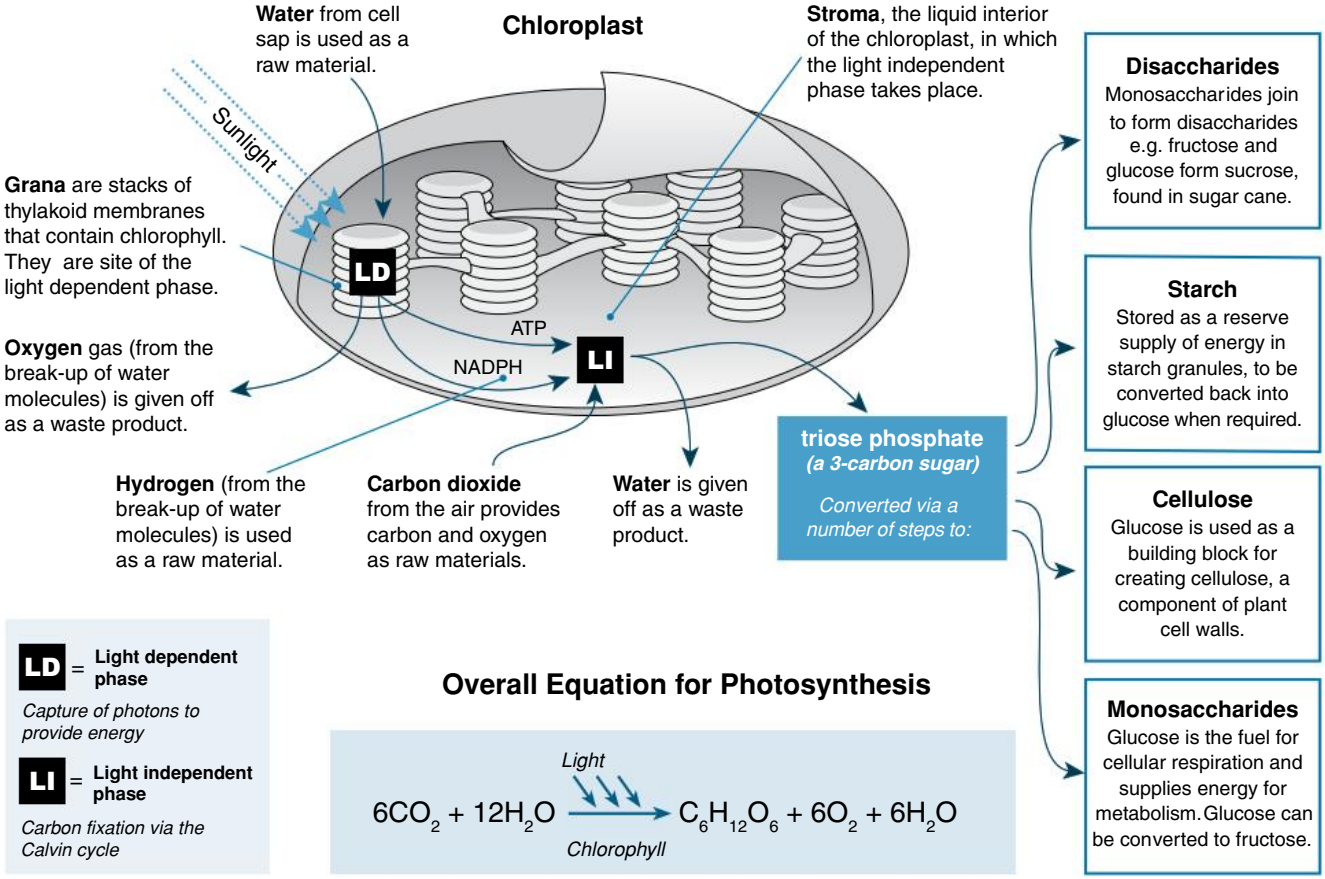
(d) Describe one improvement you could make to the experiment in order to generate more reliable data:

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32 Photosynthesis

Key Idea: Photosynthesis is the process by which light energy is used to convert CO_2 and water into glucose and oxygen.

Photosynthesis is of fundamental importance to living things because it transforms sunlight energy into chemical energy stored in molecules, releases free oxygen gas, and absorbs carbon dioxide (a waste product of cellular metabolism). Photosynthetic organisms use special pigments, called **chlorophylls**, to absorb light of specific wavelengths and capture the light energy. Like cellular respiration, photosynthesis is a redox process, but in photosynthesis, water is split and electrons are transferred together with hydrogen ions from water to CO_2, reducing it to sugar.



1. Distinguish between the two different regions of a chloroplast and describe the biochemical processes that occur in each:

   (a) _____

   _____

   _____

   (b) _____

   _____

   _____

2. State the origin and fate of the following molecules involved in photosynthesis:

   (a) Carbon dioxide: _____

   (b) Oxygen: _____

   (c) Hydrogen: _____

3. Discuss the potential uses for the end products of photosynthesis: _____

   _____

   _____

   _____

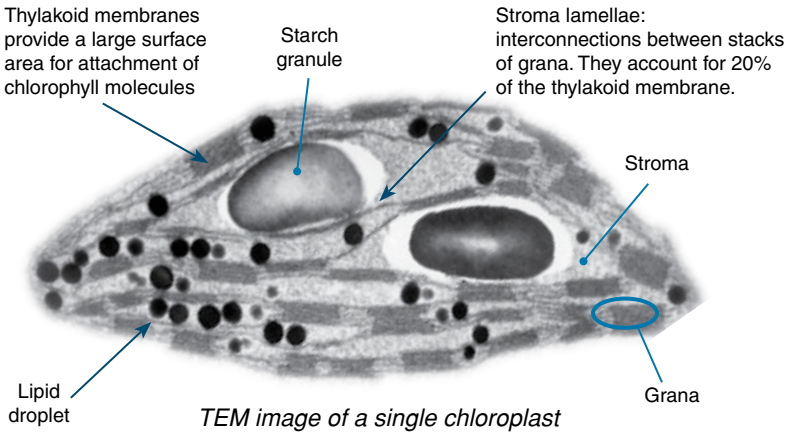
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33 Chloroplasts

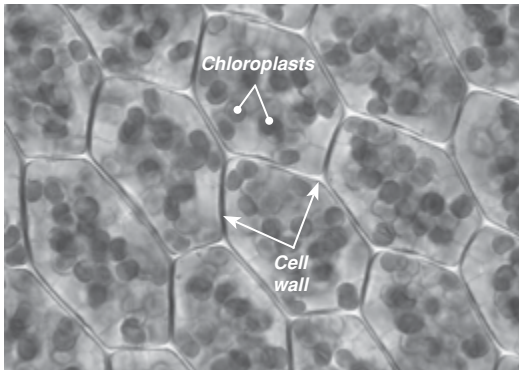
Key Idea: Chloroplasts have a complicated internal membrane structure that provides the sites for the light dependent reactions of photosynthesis.

Chloroplasts are specialised plastids in which photosynthesis occurs. A mesophyll leaf cell contains between 50-100 chloroplasts. The chloroplasts are generally aligned so that their broad surface runs parallel to the cell wall to maximise the surface area available for light absorption. Chloroplasts have an internal structure characterised by a system of membranous structures called **thylakoids** arranged into stacks called **grana**. Special pigments, called **chlorophylls** and **carotenoids**, are bound to the membranes as part of light-capturing photosystems. They absorb light of specific wavelengths and thereby capture the light energy.

The Structure of a Chloroplast



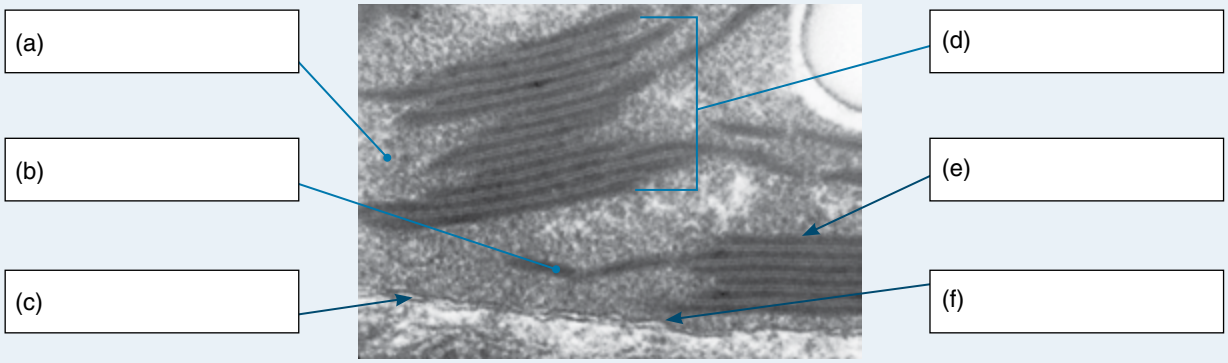
TEM image of a single chloroplast (4-6 µm diameter)



Kristian Peters

Chloroplasts visible in plant cells

1. Label the transmission electron microscope image of a chloroplast below:



2. (a) Describe where chlorophyll is found in a chloroplast: _____

_____

(b) Explain why chlorophyll is found there: _____

_____

_____

3. Explain how the internal structure of chloroplasts helps absorb the maximum amount of light: _____

_____

_____

_____

4. Explain why plant leaves appear green: _____

_____

_____

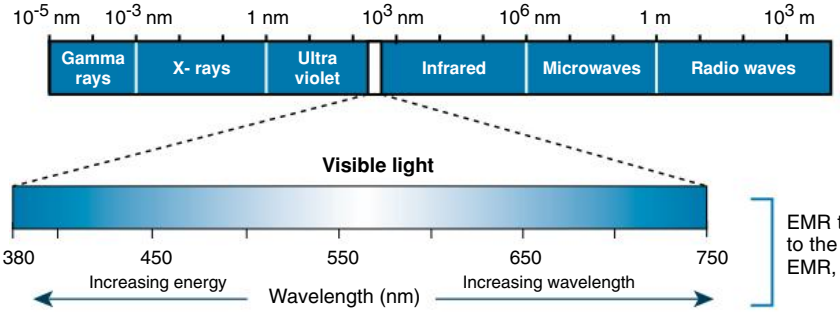
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34 Pigments and Light Absorption

Key Idea: Chlorophyll pigments absorb light of specific wavelengths and capture light energy for photosynthesis.

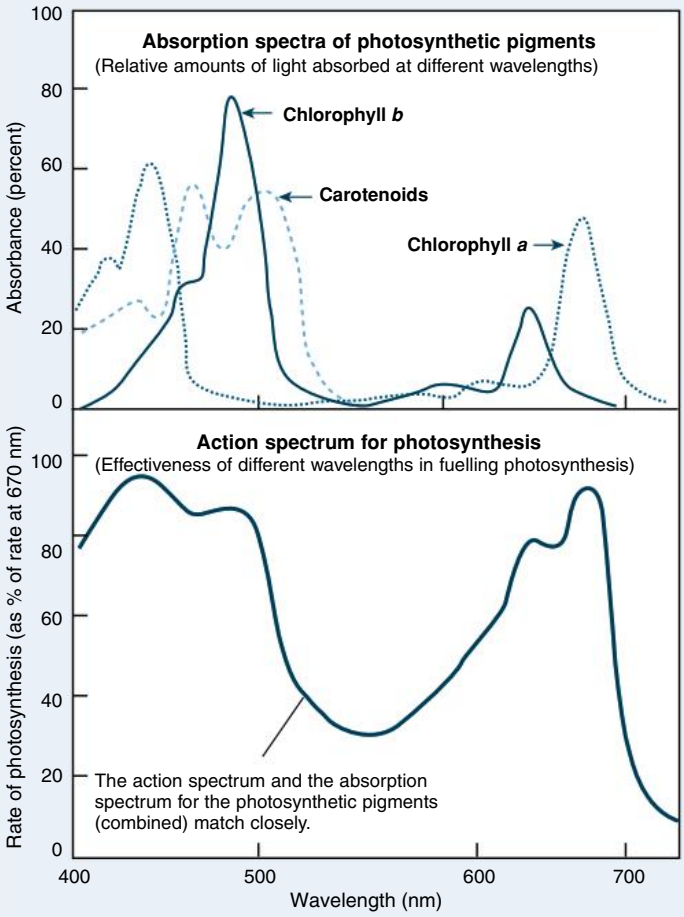
Substances that absorb visible light are called **pigments**, and different pigments absorb light of different wavelengths. The ability of a pigment to absorb particular wavelengths of light can be measured with a spectrophotometer. The light absorption vs the wavelength is called the **absorption spectrum** of that pigment. The absorption spectrum of different photosynthetic pigments provides clues to their role in photosynthesis, since light can only perform work if it is absorbed. An **action spectrum** profiles the effectiveness of different wavelengths of light in fuelling photosynthesis. It is obtained by plotting wavelength against a measure of photosynthetic rate (e.g. O_2 production).

The Electromagnetic Spectrum



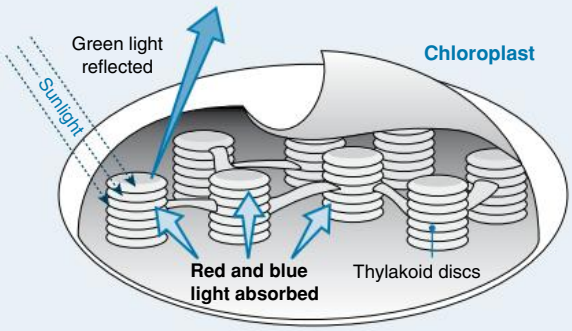
Light is a form of energy known as electromagnetic radiation (EMR). The segment of the electromagnetic spectrum most important to life is the narrow band between about 380 nm and 750 nm. This radiation is known as visible light because it is detected as colours by the human eye. It is visible light that drives photosynthesis.

EMR travels in waves, where wavelength provides a guide to the energy of the photons. The greater the wavelength of EMR, the lower the energy of the photons in that radiation.



The photosynthetic pigments of plants

The photosynthetic pigments of plants fall into two categories: **chlorophylls** (which absorb red and blue-violet light) and **carotenoids** (which absorb strongly in the blue-violet and appear orange, yellow, or red). The pigments are located on the chloroplast membranes (the thylakoids) and are associated with membrane transport systems.



The pigments of chloroplasts in higher plants (above) absorb blue and red light, and the leaves therefore appear green (which is reflected). Each photosynthetic pigment has its own characteristic absorption spectrum (top left). Only chlorophyll *a* participates directly in the light reactions of photosynthesis, but the accessory pigments (chlorophyll *b* and carotenoids) can absorb wavelengths of light that chlorophyll *a* cannot and pass the energy (photons) to chlorophyll *a*, thus broadening the spectrum that can effectively drive photosynthesis.

Left: Graphs comparing absorption spectra of photosynthetic pigments compared with the action spectrum for photosynthesis.

1. What is meant by the absorption spectrum of a pigment? _____

_____

2. Why doesn't the **action spectrum** for photosynthesis exactly match the absorption spectrum of chlorophyll *a*?

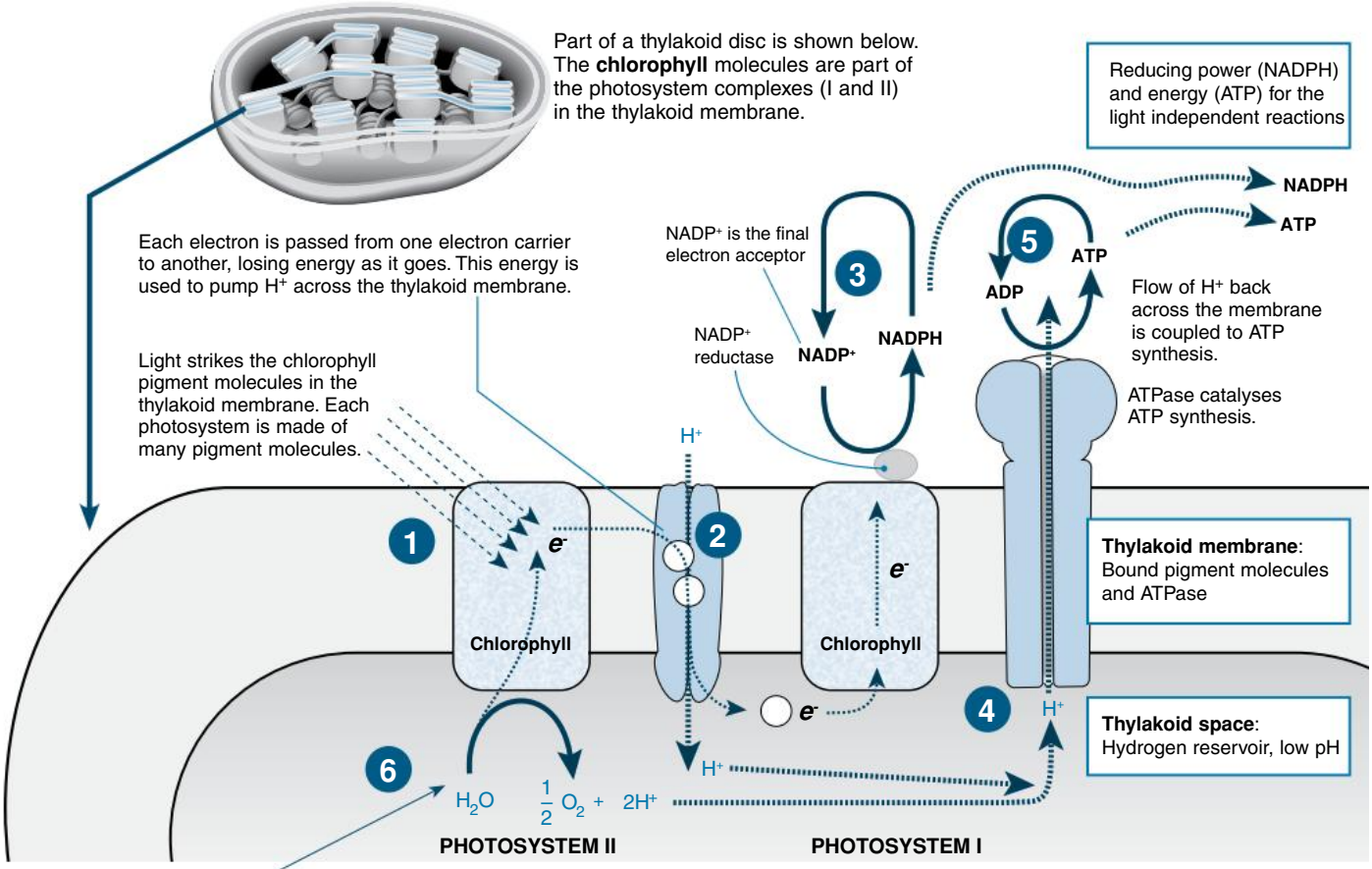
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35 Light Dependent Reactions

Key Idea: In the light dependent reactions of photosynthesis, the energy from photons of light is used to drive the reduction of $NADP^+$ and the production of ATP.

The process of photosynthesis occurs in two separate reaction systems. The light dependent reactions (the first of these) in the thylakoid of the chloroplasts capture light energy to produce **ATP** and reducing power (as **NADPH**) which is used in the light independent reactions (the second reaction system). The light dependent reactions take place in the thylakoids of the chloroplast (enlarged below).



Photolysis of water releases oxygen gas, hydrogen ions and electrons.

Photosystem II absorbs light energy to elevate electrons to a moderate energy level.

Photosystem I absorbs light energy to elevate electrons to an even higher level. Its electrons are replaced by electrons from photosystem II.

1. Where do the light dependent reactions occur? _____

2. What is the role of each of the following in the light dependent reactions:

   (a) $NADP^+$ reductase: _____

   (b) ATPase (also called ATP synthase): _____

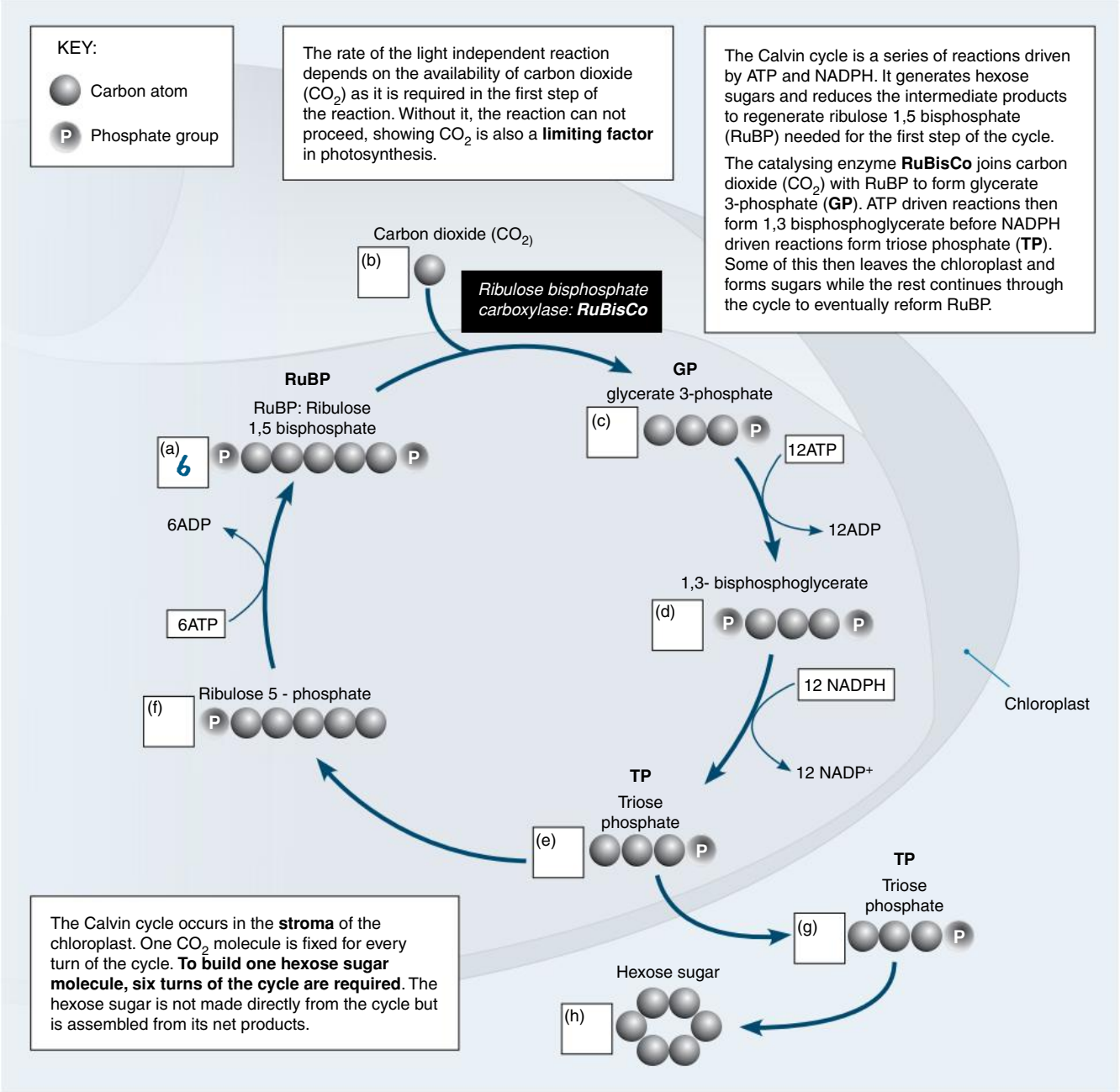
3. How are the electrons that are lost from the thylakoid system replaced? _____

4. Summarise the events of the light dependent reactions: _____

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36 Light Independent Reactions

Key Idea: The light independent reactions take place in the stroma of the chloroplast and do not require light to proceed. In the light independent reactions (or Calvin cycle) hydrogen (H^+) is added to CO_2 and a 5C intermediate to make carbohydrate. The H^+ and ATP are supplied by the light dependent reactions. In the cycle, the catalysing enzyme RuBisCo joins CO_2 to ribulose bisphosphate, a 5-carbon intermediate, which is regenerated.



1. In the boxes on the diagram above, write the number of molecules formed at each step during the formation of **one hexose sugar molecule**. The first one has been done for you:

2. Explain the importance of RuBisCo in the Calvin cycle: _____

_____

3. Identify the actual end product on the Calvin cycle: _____

4. Write the equation for the production of one hexose sugar molecule from carbon dioxide: _____

_____

5. Explain why the Calvin cycle is likely to cease in the dark for most plants, even though it is independent of light:

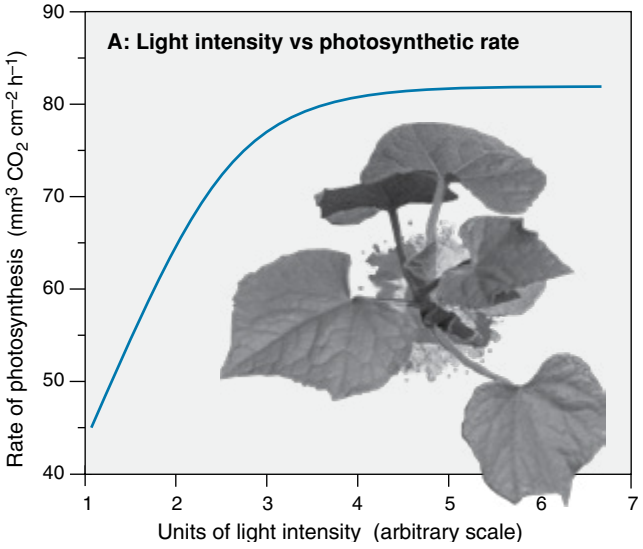
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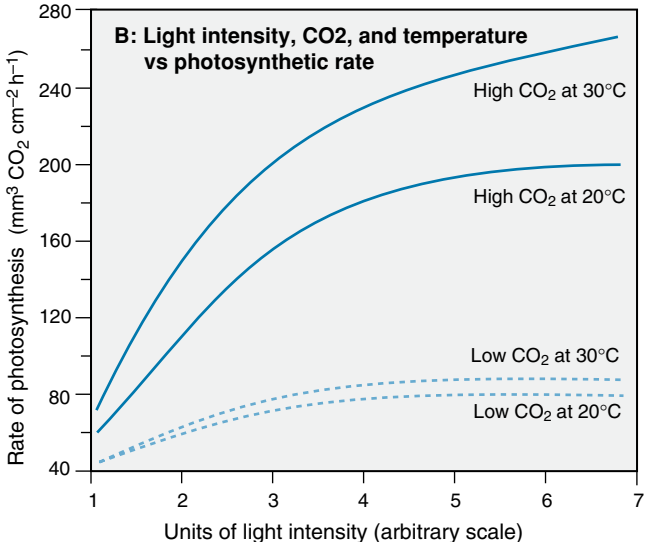
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37 Factors Affecting Photosynthesis

Key Idea: Environmental factors, such as CO_2 availability and light intensity, affect the rate of photosynthesis.

The photosynthetic rate is the rate at which plants make carbohydrate. It is dependent on environmental factors, particularly the availability of light and carbon dioxide (CO_2). Temperature is important, but its influence is less clear because it depends on the availability of the other two limiting factors (CO_2 and light) and the temperature tolerance of the plant. The relative importance of these factors can be tested experimentally by altering one of the factors while holding the others constant. The results for such an experiment are shown below.





These figures illustrate the effect of different limiting factors on the rate of photosynthesis in cucumber plants. Figure A shows the effect of different light intensities when the temperature and carbon dioxide (CO_2) level are kept constant. Figure B shows the effect of different light intensities at two temperatures and two CO_2 concentrations. In each of these experiments, either CO_2 level or temperature was changed at each light intensity in turn.

1. Based on the figures above, summarise and explain the effect of each of the following factors on photosynthetic rate:

   (a) CO_2 concentration: _____

   _____

   (b) Light intensity: _____

   _____

   (c) Temperature: _____

   _____

2. Why does photosynthetic rate decline when the CO_2 level is reduced? _____

   _____

   _____

3. (a) In figure B, explain how the effects of CO_2 concentration were distinguished from the effects of temperature:

   _____

   (b) Which factor (CO_2 or temperature) had the greatest effect on photosynthetic rate: _____

   (c) How can you tell this from the graph? _____

   _____

4. How can glasshouses be used to create an environment in which photosynthetic rates are maximised?

   _____

   _____

   _____

5. Design an experiment to demonstrate the effect of temperature on photosynthetic rate. You should include a hypothesis, list of equipment, and methods. Staple your experiment to this page.

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38 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its correct definition, as identified by its preceding letter code.

absorption spectrum

accessory pigments

acetyl coA

action spectrum

ATP

Calvin cycle

cellular respiration

chemiosmosis

chlorophyll

cristae

electron transport chain

ethanol

fermentation

glycolysis

grana

Krebs cycle

lactic acid

light dependent phase

matrix

NAD/NADP

oxidative phosphorylation

photolysis

photosynthesis

ribulose bisphosphate

stroma

thylakoid discs

A The process by which the synthesis of ATP is coupled to electron transport and the movement of protons.

B The anaerobic breakdown of glucose into two molecules of pyruvate.

C Membrane-bound compartments in chloroplasts. They are the site of the light dependent reactions of photosynthesis.

D The phase in photosynthesis where chemical energy is used for the synthesis of carbohydrate. Also called the light independent phase.

E The liquid interior of the chloroplast where the light independent phase takes place.

F The phase in photosynthesis when light energy is converted to chemical energy.

G The term to describe the light absorption vs the wavelength of a pigment.

H The end product of alcoholic fermentation.

I A nucleotide derivative, which acts as the cell's energy carrier.

J The catabolic process in which the chemical energy in complex organic molecules is coupled to ATP production.

K A 5-carbon molecule which acts as the primary CO_2 acceptor in photosynthesis.

L The stacks of thylakoids within the chloroplasts of plants.

M An anaerobic process in which pyruvate is converted to lactic acid or to ethanol and carbon dioxide.

N The stage in cellular respiration in which a derivative of pyruvate is broken down to carbon dioxide with the release of hydrogen ions.

O The process by which electrons are transferred in a series of redox reactions to a final electron acceptor such as oxygen. The energy released is used to form ATP.

P The name for the inner membranes of the mitochondria.

Q The green, membrane-bound pigment involved in the light dependent reactions of photosynthesis.

R Plant pigments that absorb wavelengths of light that chlorophyll a does not absorb.

S The inner region of the mitochondrion enclosed by the inner mitochondrial membrane.

T A chemical reaction in which the energy from photons is used to break down a chemical compound (e.g. water).

U The biochemical process that uses light energy to convert carbon dioxide and water into glucose molecules and oxygen.

V The end product of anaerobic metabolism in mammalian muscle.

W A series of electron transport proteins in the membrane of mitochondria which generate a protein gradient which can be used to generate ATP.

X Hydrogen carriers used in metabolic reactions where reducing power is needed.

Y An intermediary molecule, which transfers carbon atoms within the acetyl group to the Krebs cycle to be oxidised for energy production.

Z A profile of the effectiveness of different wavelengths of light in fuelling photosynthesis.

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39 Molecules of Life Synoptic Question

1. Enzymes are essential for catalysing biological reactions.

   (a) Describe the induced fit model of enzyme action: _____

   (b) Explain the role of cofactors and inhibitors in regulating enzyme activity: _____

2. Aerobic respiration is essential for providing energy for plant and animal cells. Discuss the process of aerobic respiration in plants and animals (you may use extra paper if required). Your discussion should include:
   - the purpose of aerobic respiration
   - the raw material(s) for aerobic respiration and the final product(s)
   - the location of aerobic respiration in plant and animal cells

3. Photosynthesis is a process in which plants use light to produce carbohydrate molecules. Discuss the process of photosynthesis and how its rate is affected by specific environmental factors (you may use extra paper if required). Your discussion should include:
   - the raw material(s) for photosynthesis and the final product(s)
   - the location of photosynthesis in plant cells
   - factors that affect the rate of photosynthesis

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QLD As required
VCE Unit 3 (2)
WA ATAR Unit 4
SA Theme C, O

Cell Signalling and Homeostasis

Key terms

action potential
ADH
apoptosis
autocrine signalling
auxin
biological transducer
blood glucose
chemoreceptor
cyclic AMP
diabetes mellitus
effector
endocrine signalling
glucagon
homeostasis
hormone
insulin
mechanoreceptor
negative feedback
nervous system
neurohormone
neurotransmitter
paracrine signalling
pheromone
photoperiodism
photoreceptors
plant growth regulator
positive feedback
receptor
response
second messenger
signal cascade
signal transduction
stimulus (pl. stimuli)
stimulus modality
synapse
target tissue
thermoregulation

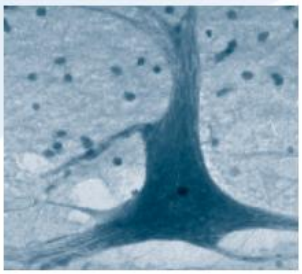


Principles of homeostasis

Learning aims and skills

Activity number

- ☐ 1 Recall the definition of homeostasis and explain the importance of maintaining a stable physiological state despite a fluctuating environment. — 40
- ☐ 2 Explain how negative feedback stabilises systems against excessive change. Using an example, such as the control of blood glucose or thermoregulation, explain the role of receptors, effectors, and negative feedback in homeostasis. — 41 42
- ☐ 3 Recognise positive feedback as a destabilising mechanism with a specific role in certain physiological processes. Describe an example of positive feedback and explain what brings the positive feedback loop to a close. — 43





Cell signalling

Learning aims and skills

Activity number

- ☐ 4 Describe and explain cell signalling, including its role in regulating the activities of organisms. Distinguish between endocrine, paracrine, and autocrine signalling. — 44
- ☐ 5 Describe signal transduction including the role of membrane receptors and second messengers (e.g. cyclic AMP) in a signal transduction pathway. Describe cell signalling mediated by a peptide (e.g. adrenaline) or a steroid. — 45 46
- ☐ 6 Explain what happens when signalling pathways are blocked or malfunction. — 47
- ☐ 7 Describe apoptosis (programmed cell death) as an example of a cellular response to specific signals. Describe the role of apoptosis in normal development and in the prevention of tumour growth. — 48 49
- ☐ 8 Describe the range of signalling molecules in plants and animals and explain how they operate. Examples include plant growth regulators such as auxin, hormones, pheromones, neurohormones, neurotransmitters, and clotting factors. — 44 50 51 55 57 61 62 63 66

Homeostatic regulation

Learning aims and skills

Activity number

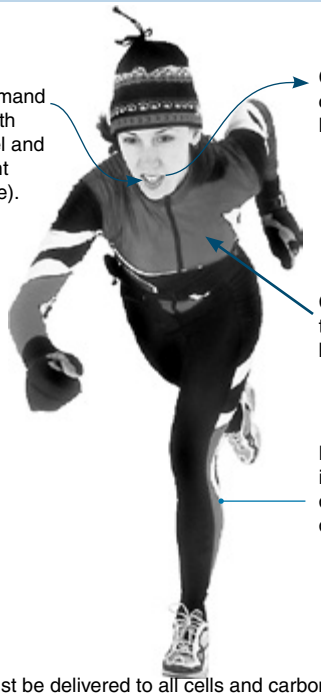
- ☐ 9 Describe the interdependence and roles of the hormonal and nervous systems in homeostatic regulation in mammals. Explain how hormones exert wide-ranging physiological effects and contrast this with neural responses. — 51 52 54
- ☐ 10 Describe examples to show your understanding of homeostatic regulation. Examples could include regulation of urine volume, regulation of blood pH or blood glucose, or thermoregulation. — 53 64-69
- ☐ 11 Describe the many homeostatic roles of blood. — 64 65 66
- ☐ 12 Describe an example to show how technology can assist in managing disorders of homeostatic regulation. — 53 70-72
- ☐ 13 Describe how the nervous system transmits information in the form of nerve impulses. Explain how sense organs act as biological transducers, identifying the stimuli to which they respond and how the stimulus modality (e.g. light) is converted to an electrical or chemical response. — 55-60

40 Maintaining Homeostasis

Key Idea: Homeostasis refers to the (relatively) constant physiological state of the body despite fluctuations in the external environment.

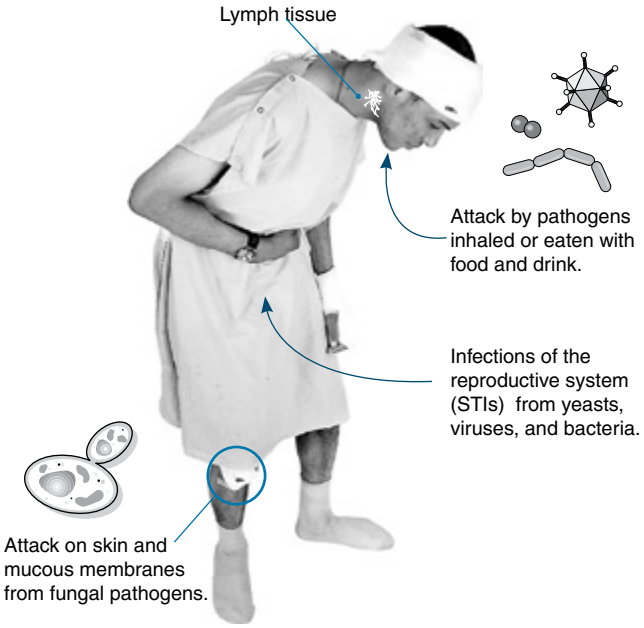
The body maintains a relatively constant physiological state, called **homeostasis**, despite fluctuations in the external environment. Homeostasis requires coordination of the hormonal (endocrine) and nervous systems, which together regulate the activity of the body's organ systems. These regulatory activities are constantly adjusted in response to stimuli from both the external and the internal environments.

Regulating Respiratory Gases



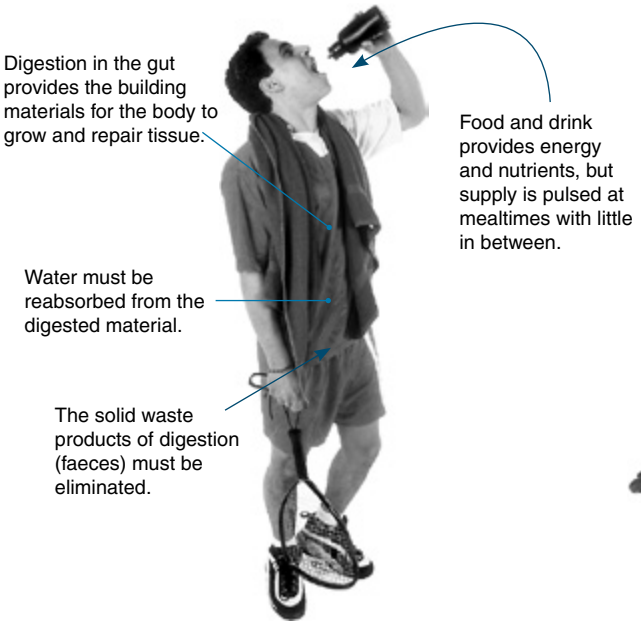
Oxygen must be delivered to all cells and carbon dioxide (a waste product of cellular respiration) must be removed. Breathing brings in oxygen and expels CO_2, and the cardiovascular and lymphatic systems circulate these respiratory gases (the oxygen mostly bound to haemoglobin). The rate of breathing is varied according to oxygen demands (as detected by CO_2 levels in the blood).

Coping with Pathogens



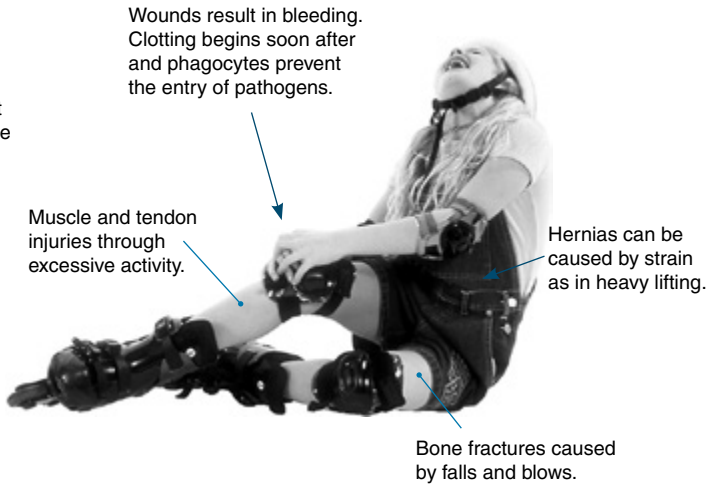
All of us are under constant attack from pathogens (disease causing organisms). The body has a number of mechanisms that help to prevent the entry of pathogens and limit the damage they cause if they do enter the body. The skin, the digestive system, and the immune system are all involved in the body's defence, while the cardiovascular and lymphatic systems circulate the cells and antimicrobial substances involved.

Maintaining Nutrient Supply



Food and drink must be taken in to maintain the body's energy supplies. The digestive system makes these nutrients available, and the cardiovascular system distributes them throughout the body. Food intake is regulated largely through nervous mechanisms while hormones control the regulation of cellular uptake of glucose.

Repairing Injuries

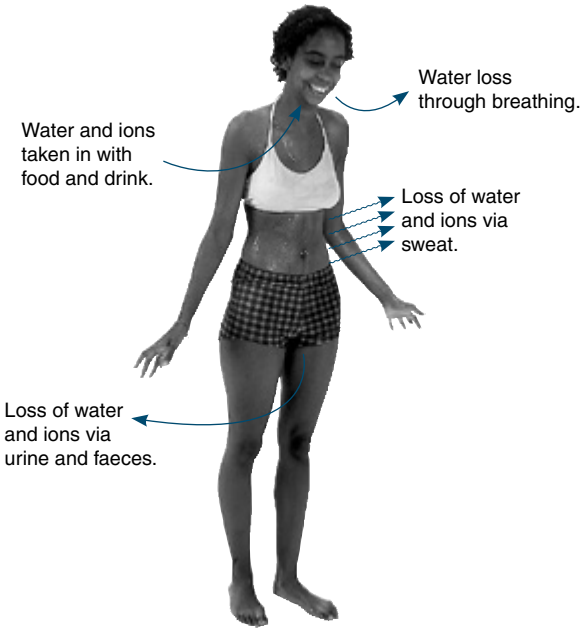


Damage to body tissues triggers the inflammatory response and white blood cells move to the injury site. The inflammatory response is started (and ended) by chemical signals (e.g. from histamine and prostaglandins) released when tissue is damaged. The cardiovascular and lymphatic systems distribute the cells and molecules involved.

KNOW
LINK 53 LINK 65 LINK 67 LINK 69 LINK 94

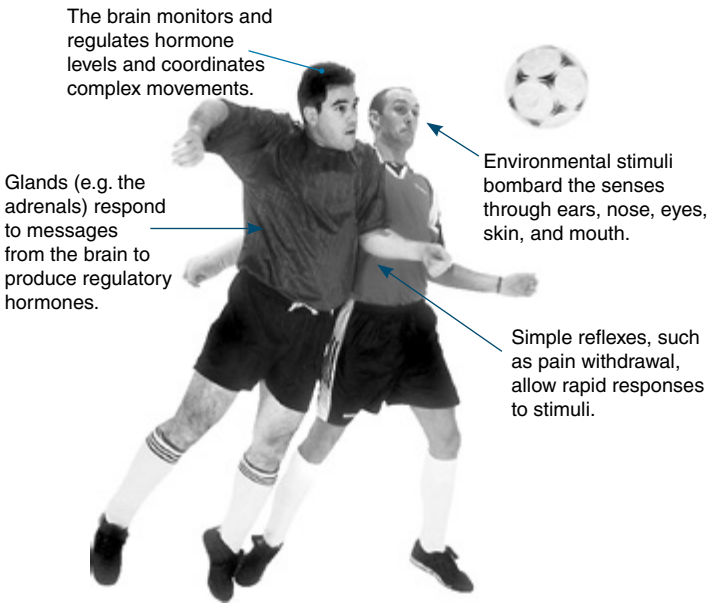
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Maintaining Fluid and Ion Balance



Fluid and electrolyte balance in the body is maintained by the kidneys (although the skin is also important). Osmoreceptors monitor blood volume and bring about the release of regulatory hormones; the kidneys regulate reabsorption of water and sodium from blood in response to levels of the hormones ADH and aldosterone. The cardiovascular and lymphatic system distribute fluids around the body.

Coordinating Responses



The body is constantly bombarded by stimuli from the environment. The brain sorts these stimuli into those that require a response and those that do not. Responses are coordinated via nervous or hormonal controls. Simple nervous responses (reflexes) act quickly. Hormones, which are distributed by the cardiovascular and lymphatic systems, take longer to produce a response and the response is more prolonged.

1. Describe two mechanisms that operate to restore homeostasis after infection by a pathogen:

   (a) _____

   _____

   (b) _____

   _____

2. Describe two mechanisms by which responses to stimuli are brought about and coordinated:

   (a) _____

   _____

   (b) _____

   _____

3. Explain two ways in which water and ion balance are maintained. Name the organ(s) and any hormones involved:

   (a) _____

   _____

   (b) _____

   _____

4. Explain two ways in which the body regulates its respiratory gases during exercise:

   (a) _____

   _____

   (b) _____

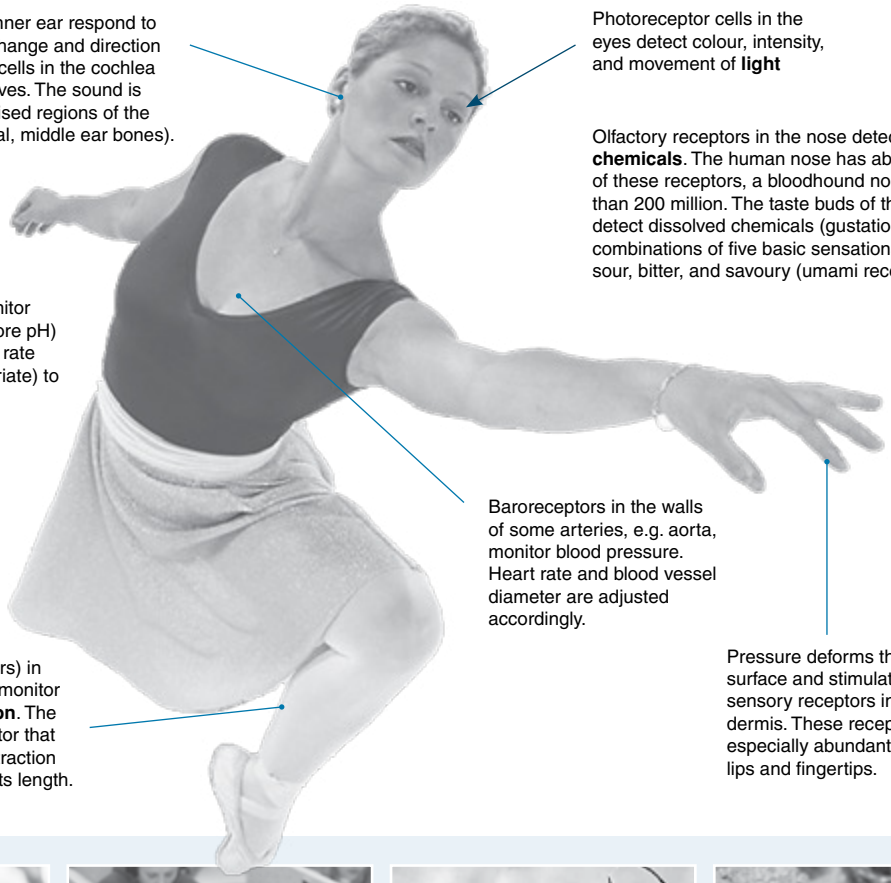
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41 Detecting Changing States

Key Idea: Sensory receptors allow the body to respond to a range of stimuli in the internal and external environments.

A **stimulus** is any physical or chemical change in the environment capable of provoking a response in an organism. Organisms respond to stimuli in order to survive. Stimuli may be either external (outside the organism) or internal (within its body). Some of the sensory receptors that animals (including humans) use to detect stimuli are shown below. Sensory receptors respond only to specific stimuli, so the sense organs an animal has determines how it perceives the world.



Hair cells in the vestibule of the inner ear respond to **gravity** by detecting the rate of change and direction of the head and body. Other hair cells in the cochlea of the inner ear detect **sound** waves. The sound is directed and amplified by specialised regions of the outer and middle ear (pinna, canal, middle ear bones).

Photoreceptor cells in the eyes detect colour, intensity, and movement of **light**

Olfactory receptors in the nose detect airborne **chemicals**. The human nose has about 5 million of these receptors, a bloodhound nose has more than 200 million. The taste buds of the tongue detect dissolved chemicals (gustation). Tastes are combinations of five basic sensations: sweet, salt, sour, bitter, and savoury (umami receptor).

Chemoreceptors in certain blood vessels, e.g. carotid arteries, monitor carbon dioxide levels (and therefore pH) of the blood. Breathing and heart rate increase or decrease (as appropriate) to adjust blood composition.

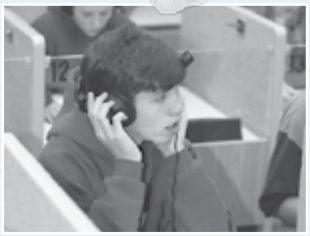
Baroreceptors in the walls of some arteries, e.g. aorta, monitor blood pressure. Heart rate and blood vessel diameter are adjusted accordingly.

Pressure deforms the skin surface and stimulates sensory receptors in the dermis. These receptors are especially abundant on the lips and fingertips.

Proprioreceptors (stretch receptors) in the muscles, tendons, and joints monitor limb position, **stretch**, and **tension**. The muscle spindle is a stretch receptor that monitors the state of muscle contraction and enables muscle to maintain its length.



Pain and temperature are detected by simple nerve endings in the skin. Deep tissue injury is sometimes felt on the skin as referred pain.



Humans rely heavily on their hearing when learning to communicate; without it, speech and language development are more difficult.



The vibration receptors in the limbs of arthropods are sensitive to movement: either sound or vibration (as caused by struggling prey).



The chemosensory Jacobson's organ in the roof of the mouth of reptiles (e.g. snakes) enables them to detect chemical stimuli.



Breathing and heart rates are regulated in response to sensory input from chemoreceptors.



Baroreceptors and osmoreceptors act together to keep blood pressure and volume within narrow limits.



Many insects, such as these ants, rely on chemical sense for location of food and communication.



Jacobson's organ is also present in mammals and is used to detect sexual receptivity in potential mates.

1. What is a stimulus and how are stimuli perceived by an organism? _____

_____

2. (a) Name one external stimulus and its sensory receptor: _____

   (b) Name one internal stimulus and its sensory receptor: _____

KNOW LINK 50 LINK 59 LINK 60 LINK 65

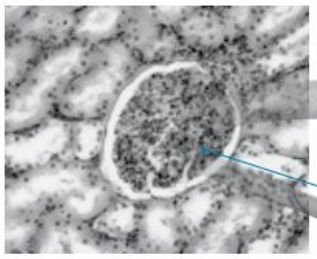
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42 Negative Feedback

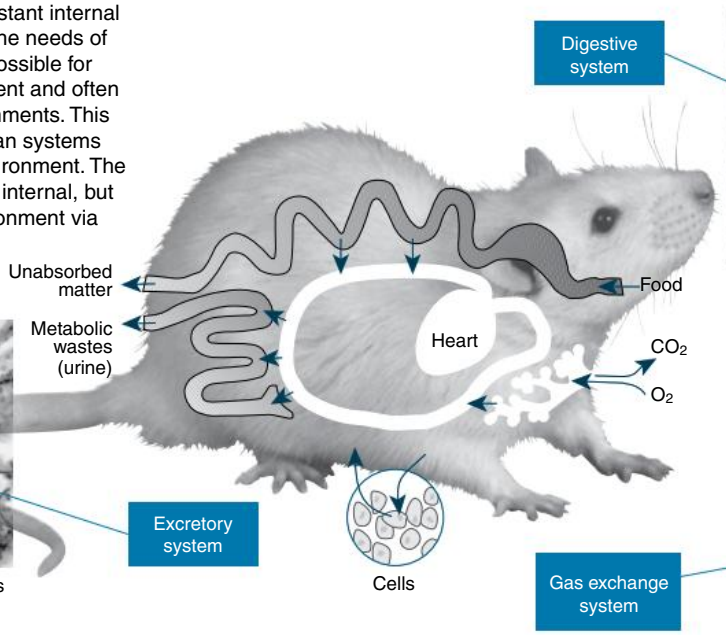
Key Idea: Negative feedback mechanisms detect changes in the internal environment away from the normal and act to return the internal environment back to a steady state.

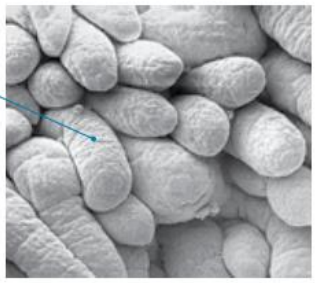
Homeostatic control systems have three functional components: a receptor to detect change, a control centre, and an effector to direct an appropriate response. In negative feedback systems, movement away from a steady state is detected and triggers a mechanism to counteract that change. **Negative feedback** has a stabilising effect, dampening variations from a set point and returning internal conditions to a steady state. Most physiological systems achieve homeostasis through negative feedback.

Organ systems maintain a constant internal environment that provides for the needs of all the body's cells, making it possible for animals to move through different and often highly variable external environments. This representation shows how organ systems permit exchanges with the environment. The exchange surfaces are usually internal, but may be connected to the environment via openings on the body surface.

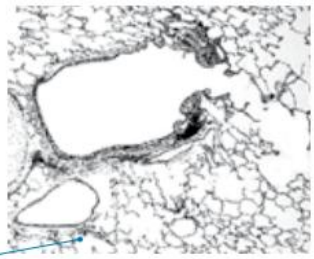


Kidney tubules exchange chemicals with the blood through capillaries





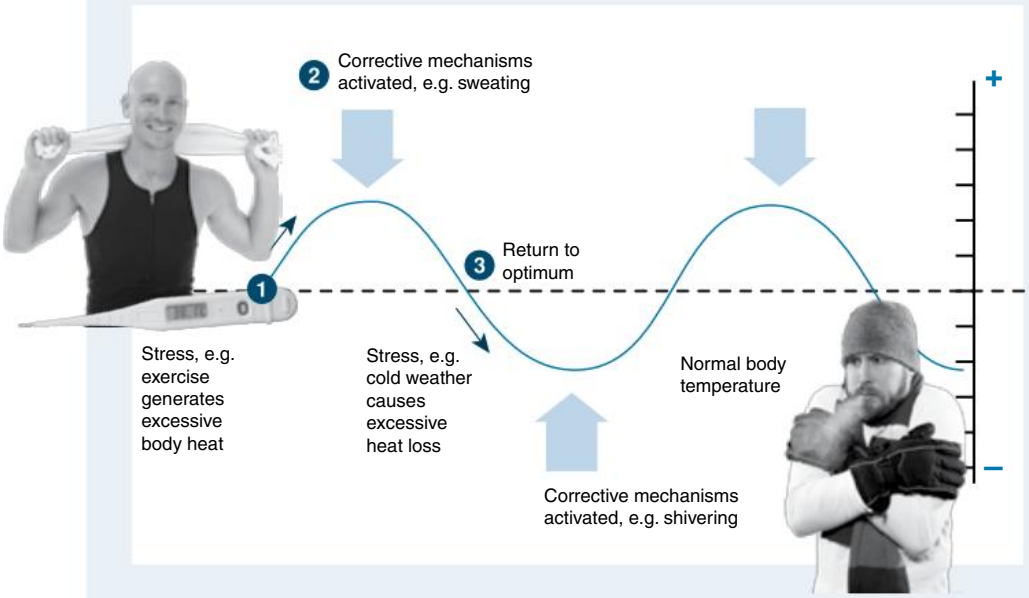
The villi of the small intestine increase surface area for nutrient absorption.



Lung tissue provides an expansive, moist surface for gas exchange.

All photos on this page: EII

Negative Feedback and Control Systems



1. A stressor, e.g. exercise, takes the internal environment away from optimum.
2. Stress is detected by receptors and corrective mechanisms (e.g. sweating or shivering) are activated.
3. Corrective mechanisms act to restore optimum conditions.

Negative feedback acts to counteract departures from steady state. The diagram shows how stress is counteracted in the case of body temperature.

1. What are the three main components of a regulatory control system in the human body and what is the role of each?

2. How do negative feedback mechanisms maintain homeostasis in a variable environment?

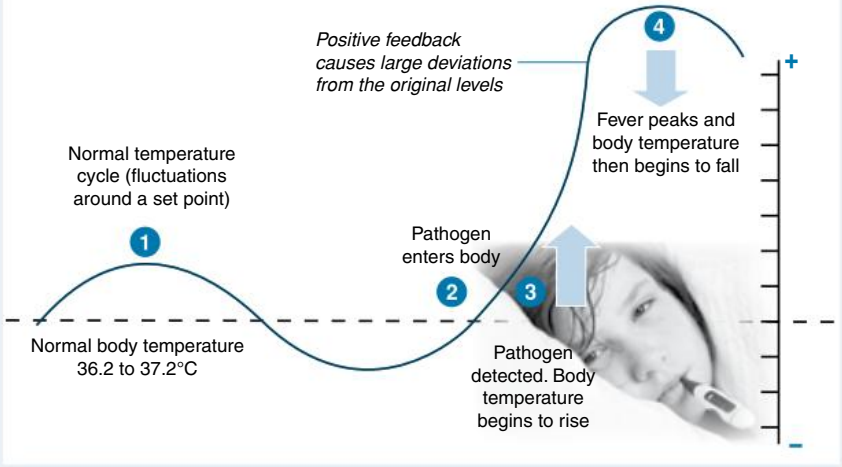
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43 Positive Feedback

Key Idea: Positive feedback results in the escalation of a response to a stimulus. It causes system instability and occurs when a particular outcome or resolution is required.

Positive feedback mechanisms amplify a physiological response in order to achieve a particular result. Labour, fever, blood clotting, and fruit ripening all involve positive feedback. Normally, a positive feedback loop is ended when the natural resolution is reached (e.g. baby is born, pathogen is destroyed). Positive feedback is relatively rare because such mechanisms are unstable and potentially damaging.

Fever, Positive Feedback and Response Escalation



1. Body temperature fluctuates on a normal, regular basis around a narrow set point.
2. Pathogen enters the body.
3. The body detects the pathogen and macrophages attack it. Macrophages release interleukins which stimulate the hypothalamus to increase prostaglandin production and reset the body's thermostat to a higher 'fever' level by shivering (the chill phase).
4. The fever breaks when the infection subsides. Levels of circulating interleukins (and other fever-associated chemicals) fall, and the body's thermostat is reset to normal. This ends the positive feedback escalation and normal controls resume. If the infection persists, the escalation may continue, and the fever may intensify. Body temperatures in excess of 43ºC are often fatal or result in brain damage.



Labour and lactation: During childbirth (above), the release of oxytocin intensifies the contractions of the uterus so that labour proceeds to its conclusion. The birth itself restores the system by removing the initiating stimulus. After birth, levels of the milk-production hormone prolactin increase. Suckling maintains prolactin secretion and causes the release of oxytocin, resulting in milk release. The more an infant suckles, the more these hormones are produced.



Ragesoss

Ethylene is a gaseous plant hormone involved in fruit ripening. It accelerates the ripening of fruit in its vicinity so nearby fruit also ripens, releasing more ethylene. Over-exposure to ethylene causes fruit to over-ripen (rot).

1. (a) What is the biological role of positive feedback loops? Describe an example: _____

_____

_____

_____

(b) Why is positive feedback inherently unstable (contrast with negative feedback)? _____

_____

_____

_____

(c) How is a positive feedback loop normally stopped? _____

_____

(d) Describe a situation in which this might not happen. What would be the result? _____

_____

_____

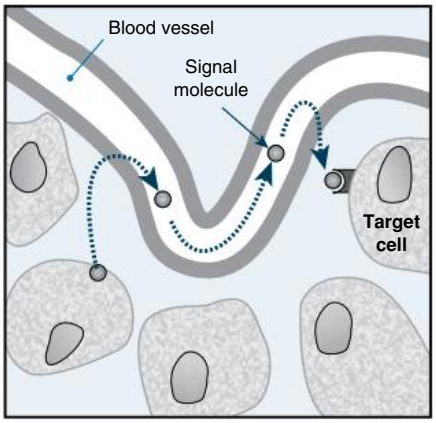
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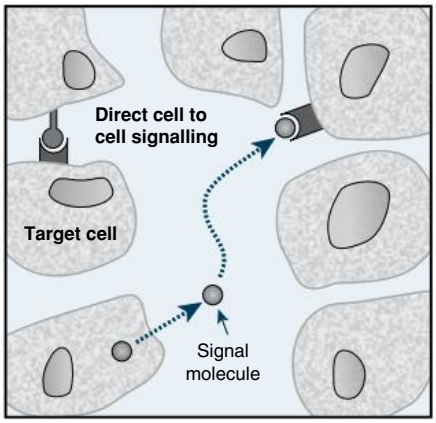
44 Types of Cell Signalling

Key Idea: Cells use signals (chemical messengers) to communicate and to gather information about, and respond to, changes in their cellular environment.

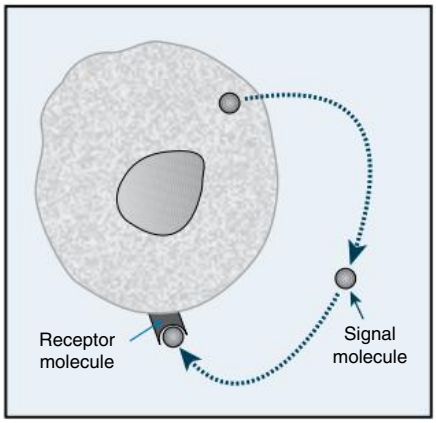
Cells communicate and bring about responses by producing and reacting to signal molecules. Three main pathways for cell signalling exist. The **endocrine** pathway involves the transport of **hormones** in the blood or haemolymph. In **paracrine** signalling, the signal travels an intermediate distance to act upon neighbouring cells. **Autocrine** signalling involves a cell producing and reacting to its own signal.



Endocrine signalling: Hormone signals are released by ductless endocrine glands and are carried by the circulatory system through the body to the target cells. Examples include sex hormones, growth factors and neurohormones such as dopamine.



Paracrine signalling: Signals released from a cell act on target cells close by. The messenger can be transferred through the extracellular fluid (e.g. at synapses) or directly between cells. Examples include histamine and prostaglandins.

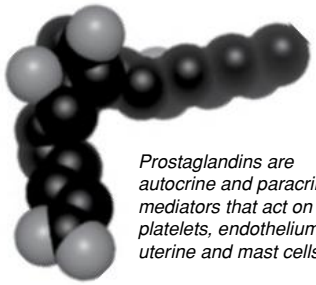


Autocrine signalling: Cells produce and react to their own signals. In vertebrates, when a foreign antibody enters the body, some T-cells produce a growth factor to stimulate their own production. The increased number of T-cells helps to fight the infection.

Signalling Receptors and Signalling Molecules



Histamine is a paracrine signal molecule involved in local immune responses.



Prostaglandins are autocrine and paracrine mediators that act on platelets, endothelium, and uterine and mast cells.

Examples of cell signalling molecules

The binding sites of cell receptors are specific only to certain **ligands** (signal molecules). This stops them reacting to every signal the cell encounters. Receptors generally fall into two main categories:

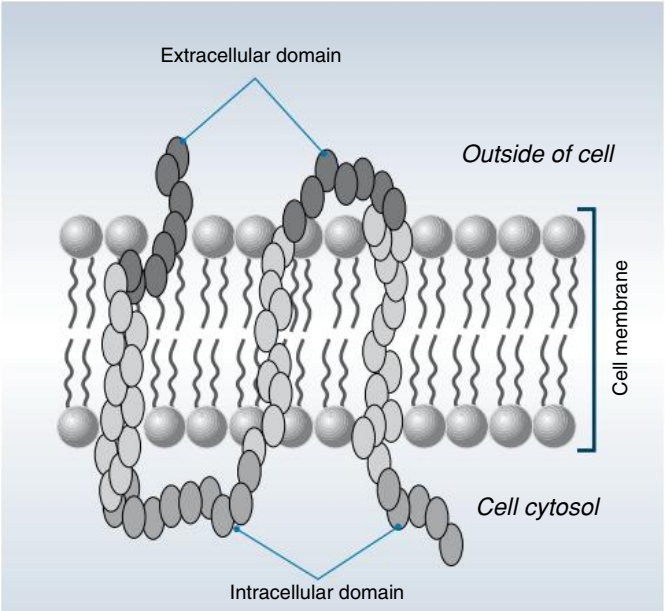
Cytoplasmic receptors

Cytoplasmic receptors, located within the cell cytoplasm, bind ligands which are able to cross the plasma membrane unaided.

Transmembrane receptors

These span the cell membrane and bind ligands which cannot cross the plasma membrane on their own. They have an extra-cellular domain outside the cell, and an intracellular domain within the cell cytosol.

Ion channels, protein kinases and G-protein linked receptors are examples of transmembrane receptors (see diagram on right).



Structure of a transmembrane receptor

1. Briefly describe the three types of cell signalling:

   (a) _____

   (b) _____

   (c) _____

2. Identify the components that all three cell signalling types have in common: _____

   _____

   _____

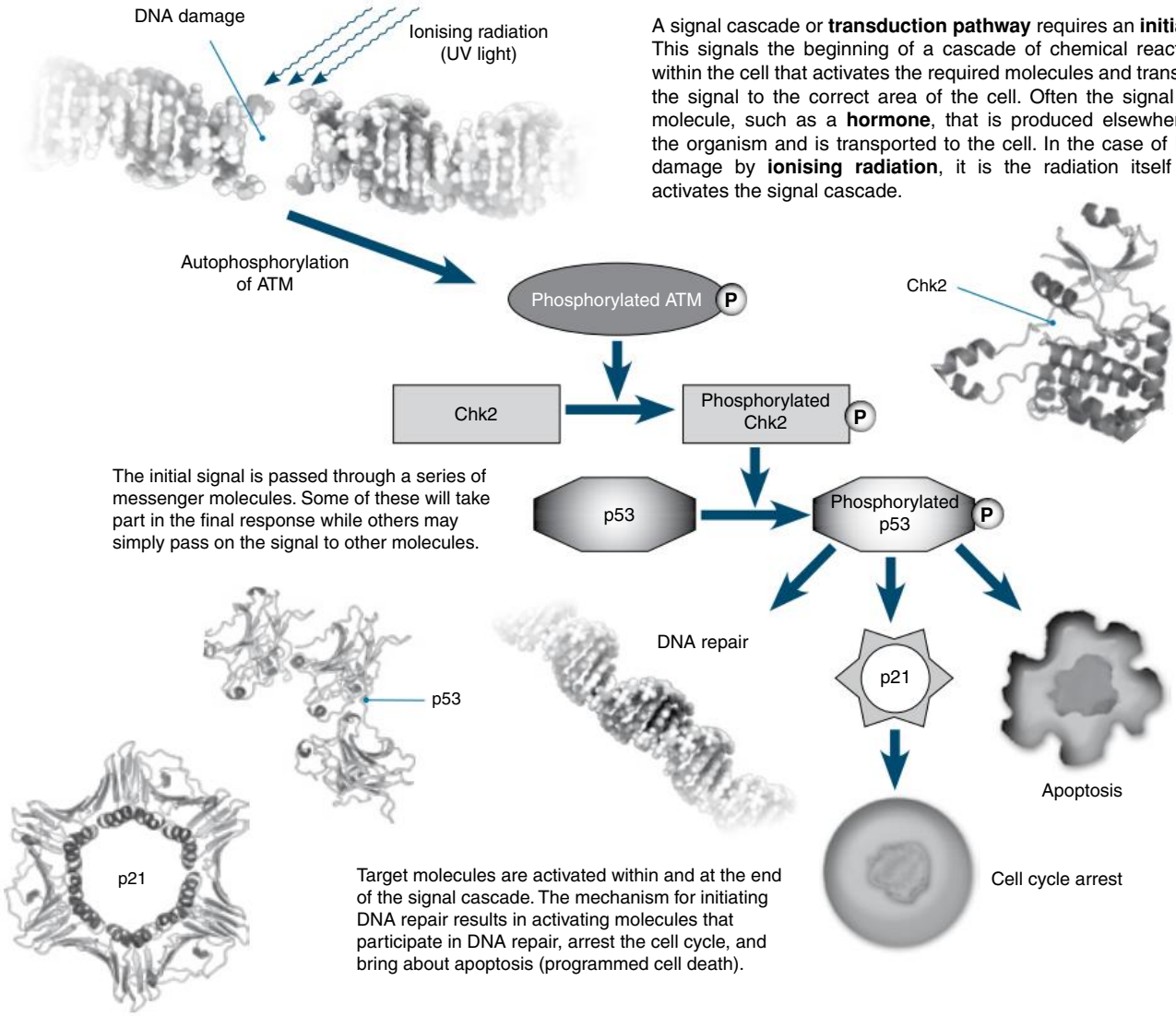
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45 Signals and Signal Cascades

Key Idea: A signal cascade occurs in response to a signal molecule and involves the activation of numerous molecules, which results in a response in the target cell.

Signals that activate processes within the cell can be external **signal molecules** or conditions that cause internal changes. These signals often cause a cascade of reactions that result in a cellular outcome. For example, the molecule adrenaline causes the breakdown of glycogen in mammals, and ionising radiation activates the protein kinase ATM, which initiates the repair of DNA. ATM is a highly conserved protein and it is a key component in DNA repair. DNA damage from ionising radiation, such as UV light, induces a signal cascade that results in repair of the DNA, arrest of cell cycle, or apoptosis (programmed cell death).

Initiating DNA Repair



A signal cascade or **transduction pathway** requires an **initiator**. This signals the beginning of a cascade of chemical reactions within the cell that activates the required molecules and transmits the signal to the correct area of the cell. Often the signal is a molecule, such as a **hormone**, that is produced elsewhere in the organism and is transported to the cell. In the case of DNA damage by **ionising radiation**, it is the radiation itself that activates the signal cascade.

The initial signal is passed through a series of messenger molecules. Some of these will take part in the final response while others may simply pass on the signal to other molecules.

Target molecules are activated within and at the end of the signal cascade. The mechanism for initiating DNA repair results in activating molecules that participate in DNA repair, arrest the cell cycle, and bring about apoptosis (programmed cell death).

1. (a) Describe how DNA repair is signalled in cells: _____

_____

_____

(b) Explain why a signal cascade can be useful in certain cell processes: _____

_____

_____

2. Use the example of DNA damage to explain why there is a strong selection pressure for the correct functioning of signal transduction pathways:

_____

_____

_____

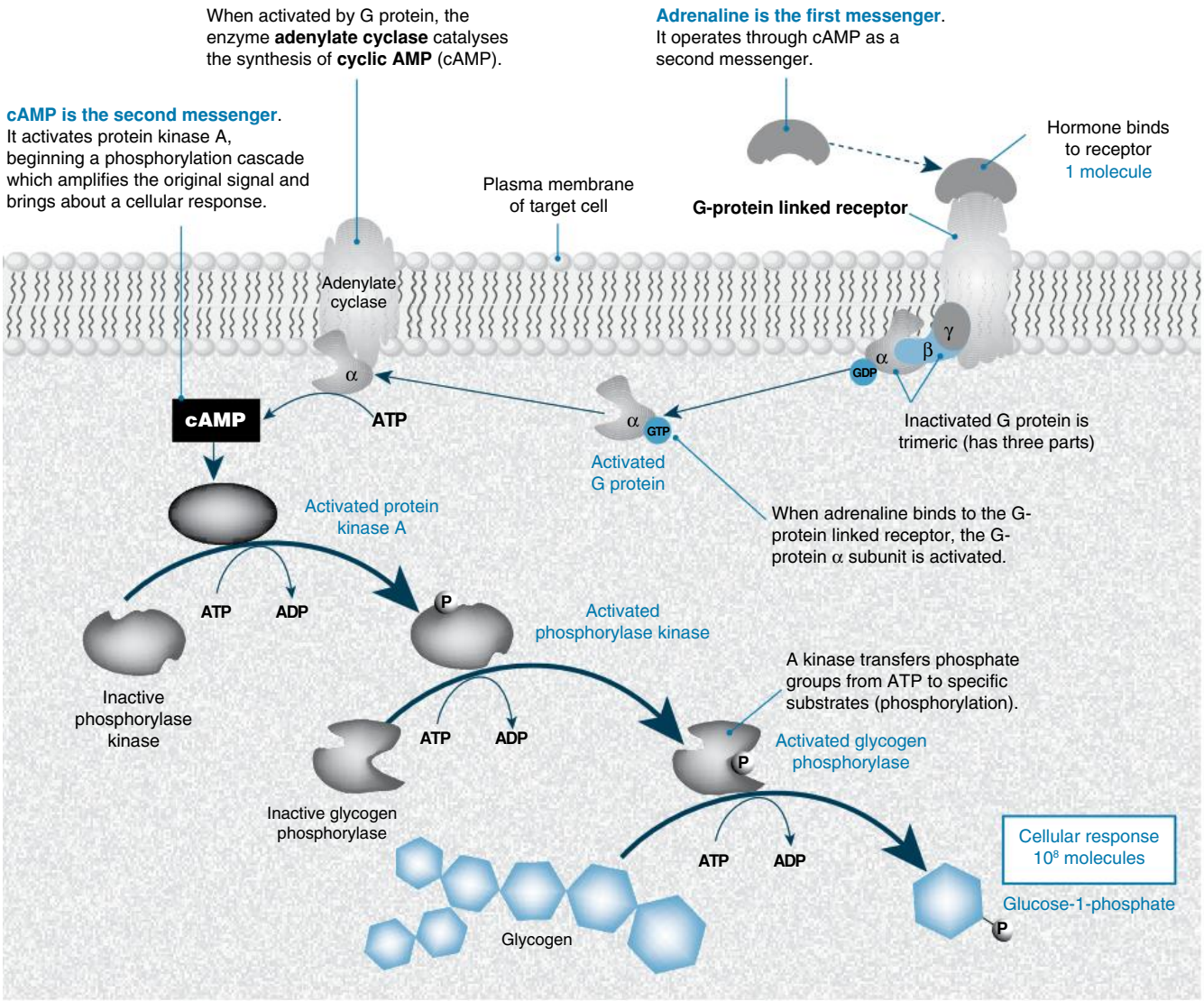
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46 Signal Transduction

Key Idea: Signal transduction is the process by which an external signal is communicated to the interior of a cell.

An extracellular signal brings about an intracellular response through a **signal transduction pathway**. The signal molecule (e.g. hormone) binds to a receptor of a target cell and starts a cascade of reactions that amplifies the original signal and brings about a response (e.g. enzyme activation). Water soluble hormones (e.g. adrenaline, below) interact with receptors in the membrane and activate a second messenger system, which links the hormone to the cellular response via a series of reactions catalysed by protein kinases. Second messengers include cyclic AMP, Ca^{2+}, and inositol triphosphate (IP3). Steroid hormones, being lipid-soluble, can enter the cell freely to interact with intracellular receptors.

Second Messenger Activation in a Signal Transduction Pathway



1. Explain why a second messenger is needed to convey a signal inside a cell from a water soluble first messenger:

2. Explain why each molecule in the cAMP signal cascade is phosphorylated:

3. What is the role of G proteins in coupling the receptor to the cellular response?

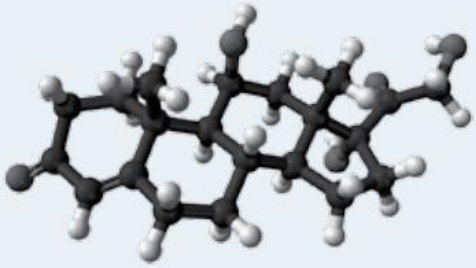
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Gene Activation by Steroid Hormones

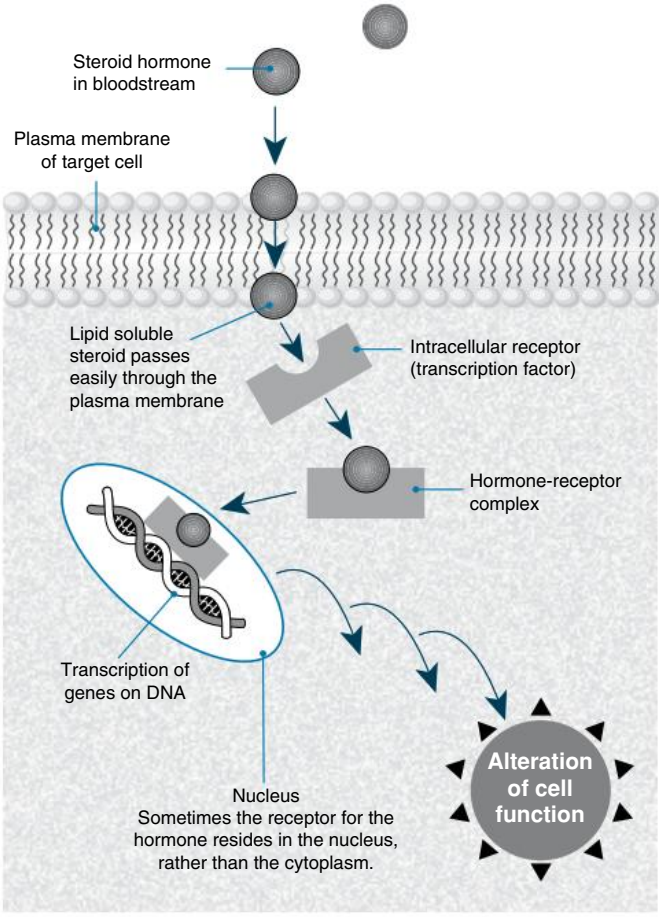
Steroid hormones are fatty molecules that act as hormones. They are involved in regulating metabolic activity, inflammation, immune function, salt and water balance, development of sexual characteristics, and response to stress and injury.

Steroid hormones are lipid soluble and diffuse freely from the blood through the plasma membrane and into the cytoplasm of target cells. In the cytoplasm, the steroid binds to a steroid receptor (**transcription factor**) to form a functional **hormone-receptor complex**. The complex is able to enter the nucleus and bind to specific DNA sequences to induce transcription of its target genes.

The best-studied steroid-binding receptors are called nuclear receptors (examples below).



Cortisol is a steroid hormone produced by the adrenal glands. Its release is triggered by ACTH from the anterior pituitary gland. Cortisol is involved in glucose metabolism and in the response to stress.



4. (a) How is a signal amplified? _____

_____

_____

(b) Why is the signal amplification important? _____

_____

5. (a) How is the action of a steroid hormone different from that of a water soluble signalling molecule?

_____

_____

(b) Suggest why steroid hormones are important in developmental processes: _____

_____

_____

6. Describe the two mechanisms by which a hormone can bring about a cellular response:

(a) _____

_____

(b) _____

_____

7. How are these two mechanisms alike? _____

_____

_____

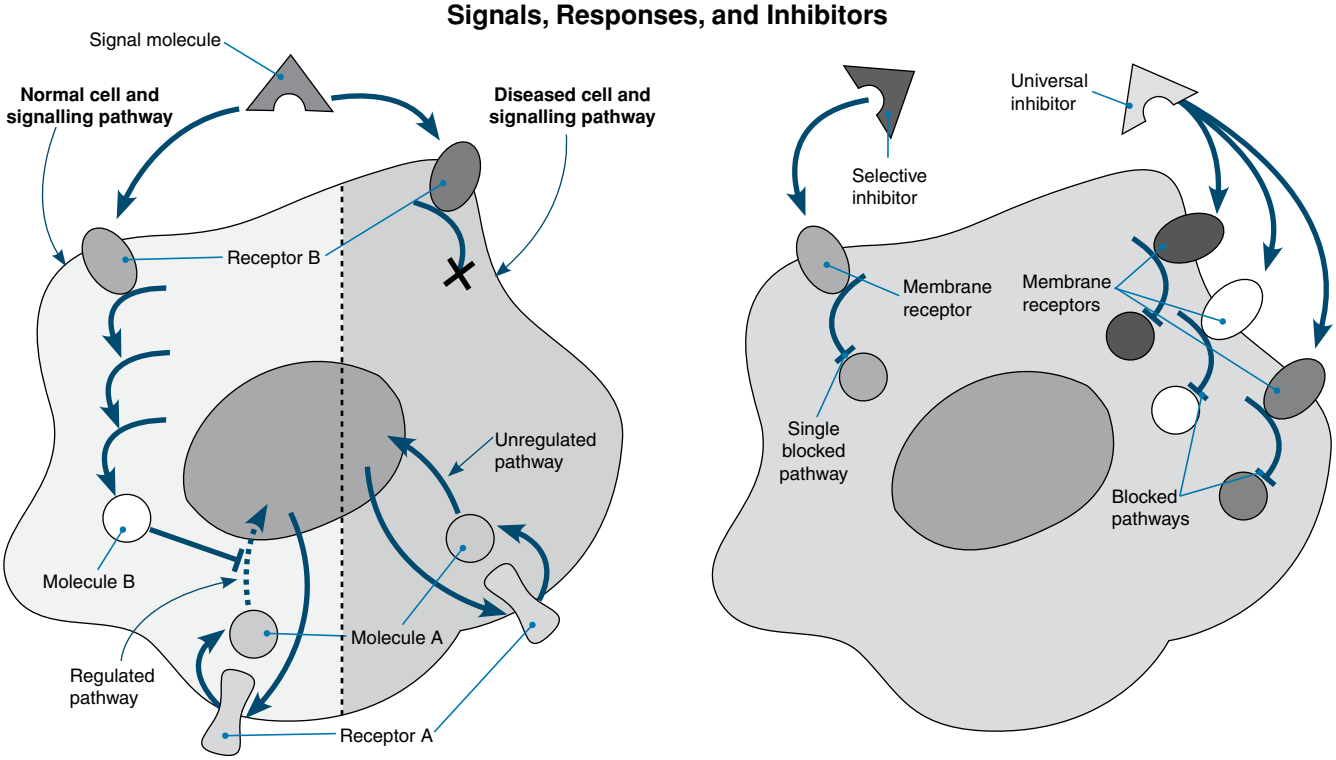
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47 Effect of Blocking Signals

Key Idea: The failure of correct cell signalling can lead to many serious diseases, including cancer.

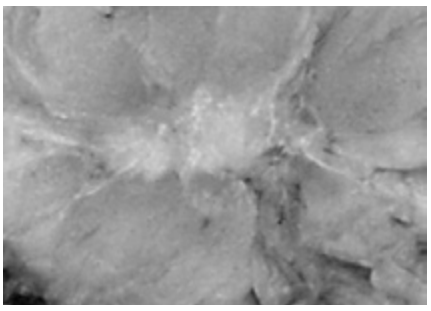
Almost as soon as biologists realised the importance of signal transduction pathways in the body, their importance in the development of disease became obvious. Failure of a pathway to operate properly would cause significant detrimental effects. Many therapeutic drugs focus on blocking the effects of overactive pathways or restoring signalling pathways when they fail. The negative effects of many toxins are caused by their actions on signal transduction pathways.

Signals, Responses, and Inhibitors

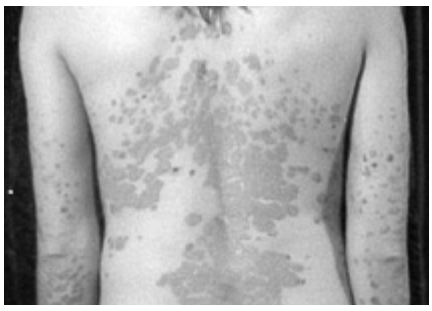


When choosing a drug to correct a transduction pathway, it is important to understand how the pathway operates. The signalling pathways of diseased cells (e.g. tumour cells) are often overactive (e.g. overexpressing receptors for growth factors). In the example above, receptor A signals the production of more receptor A via the messenger molecule A. Receptor B initiates a signal cascade activating molecule B, which inactivates molecule A and so regulates receptor A's production. The diseased cell fails to make molecule B and so cannot regulate receptor A's production.

Drug selection also involves the question of how many receptors are affected by a particular drug. Universal signal transduction inhibitors affect diseased cells by blocking many signal pathways, but also affect healthy cells, and so can cause damage outside the target area. Selective signal transduction inhibitors target a single overactive pathway, thus causing no damage outside the target area. However the specificity of these drugs may mean many different types are required to treat a disease effectively.



Breast cancer (like all cancers) is the result of cells dividing out of control. In breast cancer, the HER2 receptor protein has been identified as a major cause.



Psoriasis results from the hyper-proliferation of keratinocytes (epidermal cells) which is driven by EGFR (epidermal growth factor receptor) causing scaly patches or plaques on the skin.



Finger prick test for diabetics

Type II (adult onset) diabetes is caused by the failure of cells to respond to signalling by the hormone insulin, which results in the failure of cells to take up glucose.

1. (a) Explain the link between signal transduction and disease: _____

_____

_____

(b) Explain why understanding the link is important: _____

_____

_____

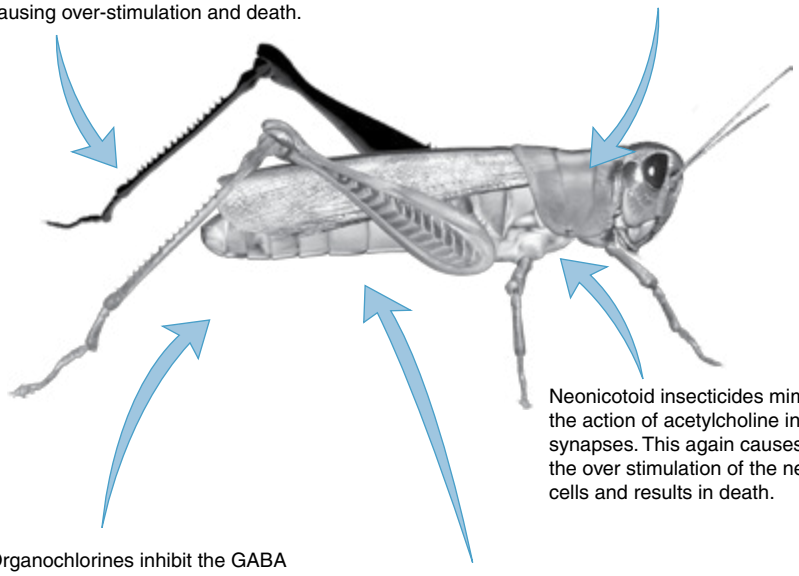
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Pesticides and Signals

Many pesticides work by affecting the signalling of nerves cells by either blocking uptake of signalling molecules or facilitating the uptake of far greater amounts than normal.

Cholinesterase inhibitors work by blocking the enzyme responsible for breaking down acetylcholine after it has transmitted a message across a synapse. As a result, the receiving neurone continues to fire, causing over-stimulation and death.

Chitin is a major part of an insect's exoskeleton. Pesticides block chitin synthesis pathway the insect from growing a new exoskeleton and moulting (so it can't grow), eventually killing the insect.



Neonicotoid insecticides mimic the action of acetylcholine in synapses. This again causes the over stimulation of the nerve cells and results in death.

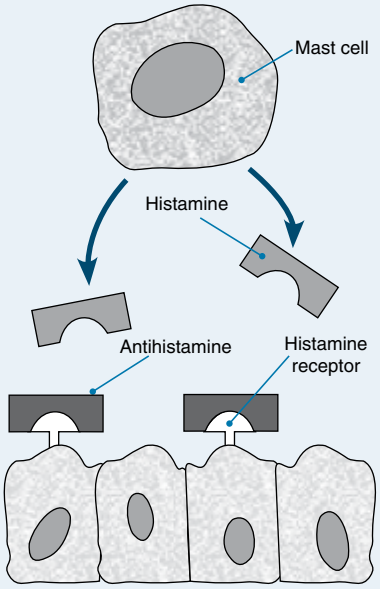
Organochlorines inhibit the GABA receptor, which itself has an inhibitory function in nerve cells. The result is over-stimulation of the nervous system and death.

Pyrethrins act on the sodium channels in neurones that pump sodium across the membrane, preventing them from closing and causing over-stimulation and death.

Action of Antihistamine

Hayfever and similar allergic reactions occur when mast cells produce the chemical histamine as part of an immune response to foreign particles such as pollen. Histamine triggers the inflammatory response and results in swelling, red eyes, and a runny nose.

Antihistamines block the histamine receptor of cells and so block the signal for the inflammatory response.



2. (a) Describe the action of a universal signal transduction inhibitor: _____

(b) Describe the action of a selective transduction inhibitor: _____

(c) Explain the role of these drugs in controlling a disease: _____

3. (a) Describe the general function of insecticides that act on the nervous system: _____

(b) Explain why pesticides based on cholinesterase inhibitors are also toxic to humans while pesticides based on chitin synthesis inhibitors are not:

4. Explain how antihistamines prevent hayfever: _____

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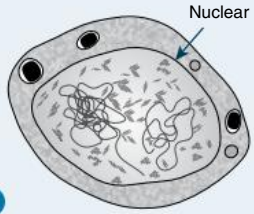
48 Apoptosis: Programmed Cell Death

Key Idea: Apoptosis is a controlled cell suicide that maintains cell numbers and sculpts body parts during development.

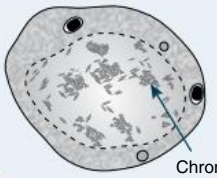
Apoptosis or programmed cell death (PCD) is a normal and necessary mechanism in multicellular organisms to trigger the death of a cell. Apoptosis helps to maintain adult cell numbers and is a defence against damaged or dangerous cells, such as virus-infected cells and cells with DNA damage. Apoptosis also has a role in "sculpting" embryonic tissue during development, e.g. in the formation of fingers and toes in a developing human embryo and resorption of the larval tail during amphibian metamorphosis. PCD involves an orderly series of events that result in set changes in cell morphology and end in cell death. The process safely disposes of cell remains and fragments. This is in contrast to **necrosis**, in which traumatic damage to the cell results in spillage of cell contents. Apoptosis is tightly regulated by a balance between the factors promoting cell survival and those triggering cell death. An imbalance between these factors leads to defective apoptotic processes and is implicated in many diseases.

Stages in Apoptosis

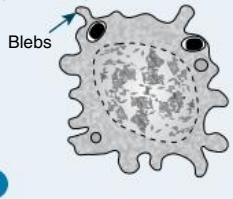
Apoptosis is a normal cell suicide process in response to cell signals. It characterised by an overall compaction (shrinking) of the cell and its nucleus, and the orderly dissection of chromatin by endonucleases. Death is finalised by phagocytosis of the dying cell. The cell contents remain membrane-bound and there is no inflammation.



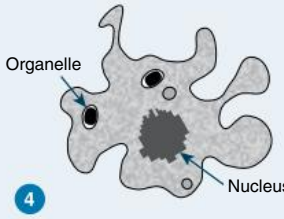
1 The cell shrinks and loses contact with neighbouring cells. The chromatin condenses and begins to degrade.



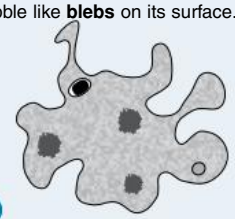
2 The nuclear membrane degrades. The cell loses volume. The chromatin clumps into **chromatin bodies**.



3 **Zeiosis**: The plasma membrane forms bubble like **blebs** on its surface.



4 The nucleus collapses, but many membrane-bound organelles are not affected.



5 The nucleus breaks up into spheres and the DNA breaks up into small fragments.



6 The cell breaks into numerous **apoptotic bodies**, which are quickly resorbed by phagocytosis.





Ed Uthman

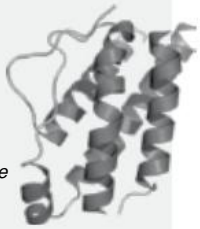
In humans, the mesoderm initially formed between the fingers and toes is removed by apoptosis. 41 days after fertilisation (top left), the digits of the hands and feet are webbed, making them look like small paddles. Apoptosis selectively destroys this superfluous webbing and, later in development, each of the digits can be seen (right).

Regulating Apoptosis

Apoptosis is a complicated and tightly controlled process, distinct from cell necrosis (uncontrolled cell death), when the cell contents are spilled. Apoptosis is regulated through both:

Positive signals, which prevent apoptosis and allow a cell to function normally. They include:

- interleukin-2
- bcl-2 protein and growth factors



Interleukin-2 is a positive signal for cell survival. Like other signalling molecules, it binds to surface receptors on the cell to regulate metabolism.

Negative signals (death activators), which trigger the changes leading to cell death. They include:

- inducer signals generated from within the cell itself in response to stress, e.g. DNA damage or cell starvation.
- signalling proteins and peptides such as lymphotoxin.

1. The photograph (right) depicts a condition called syndactyly. What might have happened during development to result in this condition?



2. Describe one difference between apoptosis and necrosis: _____

3. Describe two situations, other than digit formation in development, in which apoptosis plays a crucial role:

(a) _____

(b) _____

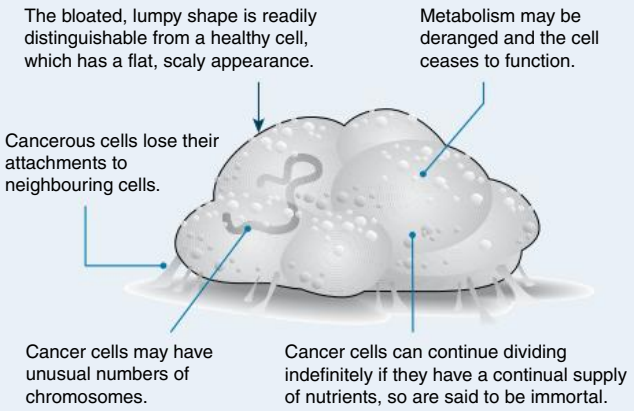
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49 Cancer

Key Idea: Cancer is characterised by the progressive and uncontrolled growth of abnormal cells caused by mutations in genes that control cell growth and replication.

Cancer is a term used for diseases in which abnormal cells divide uncontrollably, forming tissue masses (tumours) and invading other tissues. It not a new disease, nor is it restricted just to humans. Environmental, genetic, and biological factors are all implicated in contributing to cancer. The incidence of many cancers increases with age probably as a result of age-related changes in DNA repair and gene regulation.

Features of Cancer Cells

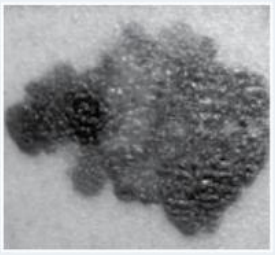


The diagram above shows a single **lung cell** that has become cancerous. It no longer carries out the role of a lung cell, and instead takes on a parasitic 'lifestyle', taking from the body what it needs in the way of nutrients and contributing nothing in return. The rate of cell division is greater than in normal cells in the same tissue because there is no resting phase between divisions.

Common Cancers

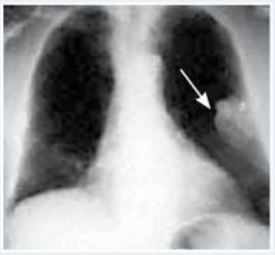
Skin cancer (**melanoma**): Cancerous tumours usually develop on skin exposed to **UV light**, but can occur anywhere on the body. Melanomas generally develop from an existing mole, which may enlarge, become lumpy, bleed, change colour, or develop a spreading black edge (as shown below). Melanomas are **malignant** and often spread to other parts of the body.

Lung cancer: Lung cancer is one of the most common of all malignant tumours and 98% of all cases are associated with cigarette smoking. Symptoms of lung cancer include coughing up blood, chest pain, breathlessness, and headache if the tumour has metastasised to the brain. These symptoms are generally caused by the tumour impairing lung function.



NCI

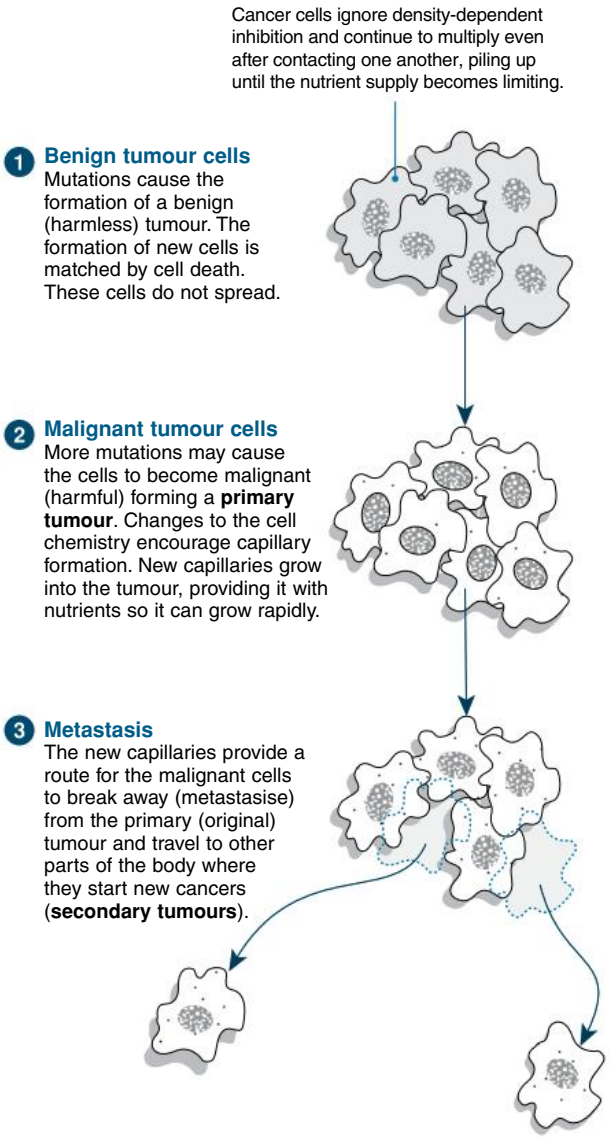
Melanoma with the characteristic spreading black edge.



Chest X-ray showing the dense mass of a tumour.

Stages in the Formation of Cancer

A tumour begins when the genes controlling cell growth and multiplication are transformed by cancer-causing agents (**carcinogens**) into cancer-causing **oncogenes**. The p53 gene, which normally prevents division of damaged cells, is the most well studied. The p53 gene is altered in 40% of all cancers. Once a cell becomes **malignant** (harmful), the change in its oncogenes is passed to all daughter cells.



1. Distinguish between benign and malignant tumours: _____

_____

_____

2. What is the genetic basis of malignancy in cells? _____

_____

3. Why is the rate of cell division in tumour cells so high? _____

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50 Pheromones

Key Idea: A pheromone is a chemical produced by an animal and released into the external environment where it affects the physiology or behaviour of members of the same species. **Pheromones**, which are often sex attractants, are common amongst insects and mammals, and commonly relate to reproductive behaviour. Many mammals, including canids and all members of the cat family, use scent marking to mark territories and advertise their readiness to mate. Most mammals also release a mammary pheromone that triggers nursing behaviour in the young. Pheromones are also used as signalling molecules in social insects such as bees, wasps, and ants. They may be used to mark a scent trail to a food source or to signal alarm. Pheromones are widely used as baits to attract and trap pests.

Pheromones and Animal Communication



Pheromones produced by a honey bee queen and her daughters, the workers, maintain the social order of the colony. The pheromone is a blend of unsaturated fatty acids.



Like mammals, reptiles detect chemicals with the vomeronasal (Jacobson's) organ. The flicking of a snake's tongue allows the snake to chemically sample its environment.



In mammals, pheromones are used to signal sexual receptivity and territorial presence, or to synchronise group behaviour. Pheromone detection relies on the vomeronasal organ (VNO), an area of receptor tissue in the nasal cavity. Mammals use a flehmen response, in which the upper lip is curled up, to better expose the VNO to the chemicals of interest.



Communication in ants and other social insects occurs through detection of pheromones. Foraging ants will leave a trail along the ground which other ants will follow and reinforce until the food source is depleted. Ants also release alarm substances, which will send other ants in the vicinity into an attack frenzy. These signals dissipate rapidly if not reinforced.





The feathery antennae of male moths are stereochemically specialised to detect the pheromone released by a female moth. Male moths can detect concentrations as low as 2ppm and will fly upwind toward the source to mate with the female. This sex attractant property of pheromones is exploited in pheromone traps, which are widely used to trap insect pests in orchards.



Photo courtesy of Cereal Research Centre, AAFC

1. (a) Distinguish between hormones and **pheromones**: _____

_____

_____

(b) What is the significance of pheromones being species specific? _____

_____

2. Giving examples, briefly describe the role of pheromones in three aspects of animal behaviour:

(a) _____

(b) _____

(c) _____

3. From what you know of pheromone activity, how would a pheromone trap operate to control an insect pest?

_____

_____

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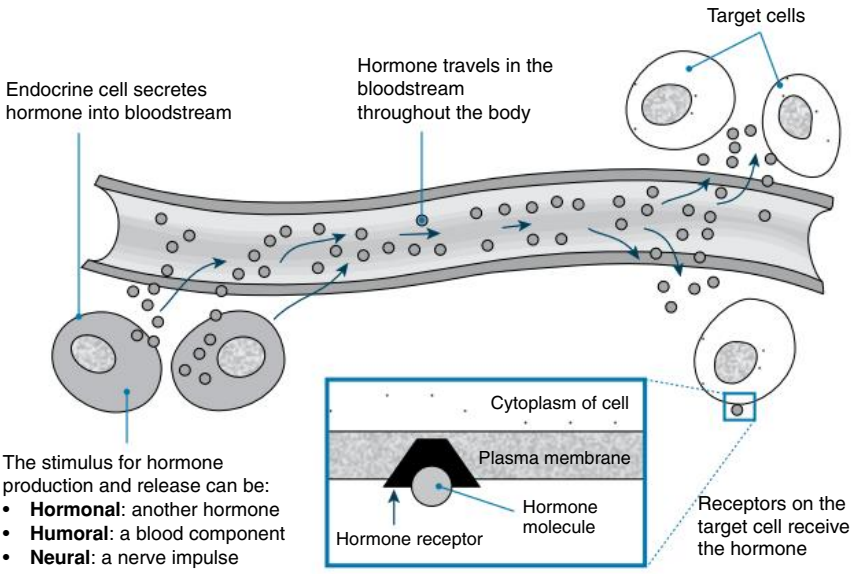
51 Hormonal Regulation

Key Idea: The endocrine system regulates physiological processes by releasing chemical messengers (hormones) into the bloodstream to interact with target cells.

The endocrine system is made up of endocrine cells (organised into endocrine glands) and the hormones they produce. Hormones are potent chemical regulators. They are produced in very small quantities but can exert a very large effect on metabolism. Endocrine glands secrete hormones directly into the bloodstream rather than through a duct or tube. The basis of hormonal control and the role of negative feedback mechanisms in regulating hormone levels are described below.

The Mechanism of Hormone Action

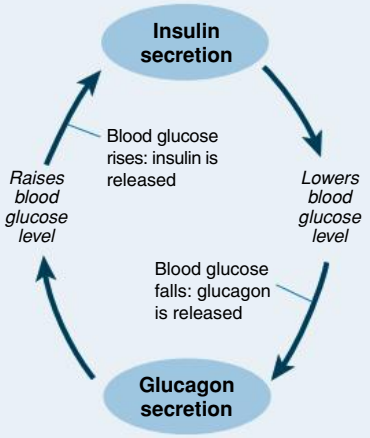
Endocrine cells produce hormones and secrete them into the bloodstream where they are distributed throughout the body. Although hormones are broadcast throughout the body, they affect only specific target cells. These target cells have receptors on the plasma membrane which recognise and bind the hormone (see inset, below right). The binding of hormone and receptor triggers the response in the target cell. Cells are unresponsive to a hormone if they do not have the appropriate receptors.



The stimulus for hormone production and release can be:

- **Hormonal**: another hormone
- **Humoral**: a blood component
- **Neural**: a nerve impulse

Antagonistic Hormones



The effects of one hormone are often counteracted by an opposing hormone. Feedback mechanisms adjust the balance of the two hormones to maintain a physiological function.
Example: insulin acts to decrease blood glucose and glucagon acts to raise it.

1. (a) What are **antagonistic hormones**? Describe an example of how two such hormones operate:

Example: _____

(b) Describe the role of feedback mechanisms in adjusting hormone levels (explain using an example if this is helpful):

2. How can a hormone influence only the target cells even though all cells may receive the hormone?

3. Explain why hormonal control differs from nervous system control with respect to the following:

(a) The speed of hormonal responses is slower: _____

(b) Hormonal responses are generally longer lasting: _____

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52 The Endocrine System

Key Idea: The endocrine system regulates specific aspects of metabolism through hormones, which are secreted from endocrine glands.

The **endocrine glands** are ductless glands distributed throughout the body. They secrete **hormones** (chemical messengers) which are carried in the blood to exert a specific effect on target cells. They are then broken down and excreted. The hypothalamus is part of the brain and not strictly an endocrine gland, but it links the nervous and endocrine systems and helps to regulate the body's activities.

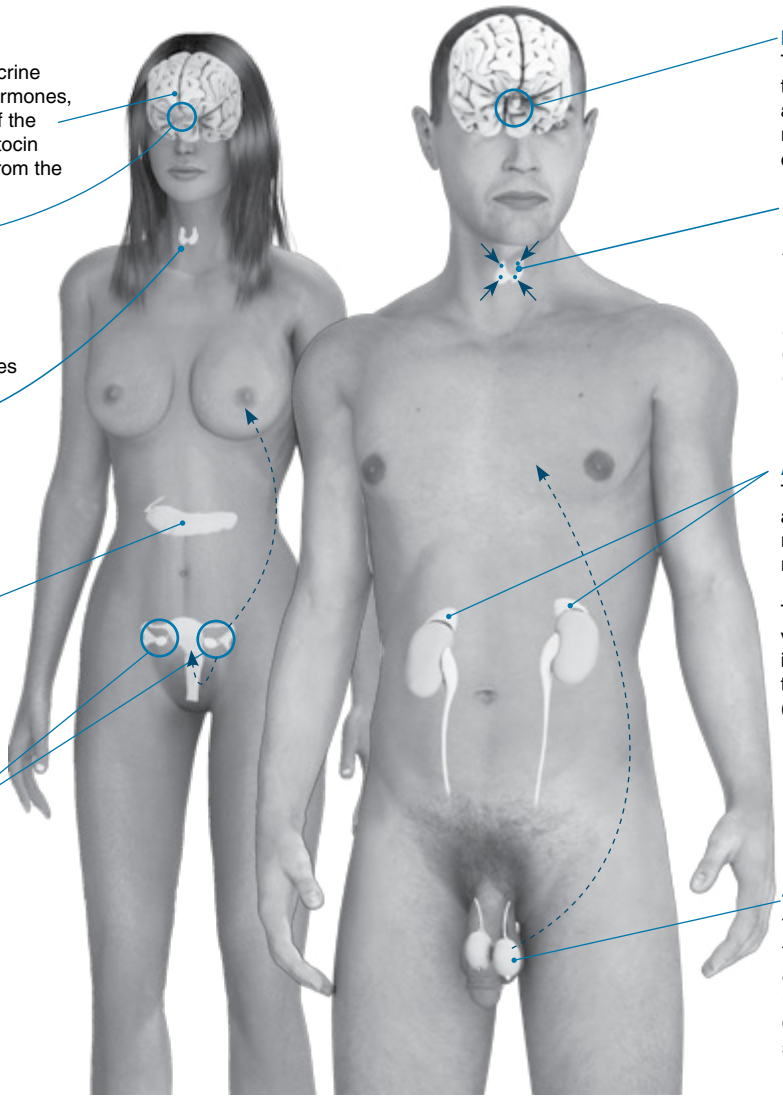
Hypothalamus
Coordinates nervous and endocrine systems. Secretes releasing hormones, which regulate the hormones of the anterior pituitary. Produces oxytocin and ADH, which are released from the posterior pituitary.

Pineal
This small gland in the brain secretes melatonin, which regulates the sleep-wake cycle. Melatonin secretion follows a circadian rhythm and coordinates reproductive hormones too.

Thyroid gland
Secretes thyroxine, an iodine containing hormone needed for normal growth and development. Thyroxine stimulates metabolism and growth via protein synthesis.

Pancreatic islets
Specialised α and β endocrine cells in the pancreas produce glucagon and insulin. Together, these control blood sugar levels.

Ovaries (in females)
The ovaries produce oestrogen and progesterone. These hormones control and maintain female characteristics, stimulate the menstrual cycle, maintain pregnancy, and prepare the mammary glands for lactation.



Pituitary gland
The pituitary is located below the hypothalamus. It secretes at least nine hormones that regulate the activities of other endocrine glands.

Parathyroid glands
Located on the surface of the thyroid, they secrete PTH (parathyroid hormone), which regulates blood calcium levels and promotes the release of calcium from bone. High levels of calcium in the blood inhibit PTH secretion.

Adrenal glands
The adrenal medulla produces adrenaline and noradrenaline responsible for the fight or flight response.

The adrenal cortex produces various steroid hormones, including cortisol (response to stress) and aldosterone (sodium regulation).

Testes (in males)
The testes of males produce testosterone, which controls and maintains "maleness" (muscular development and deeper voice), and promotes sperm production.

1. How is a hormone different from a neurotransmitter? _____

_____

2. Draw lines to connect each of the following endocrine glands with its correct role in the body:

(a) Pituitary gland	The hormone from this gland regulates the levels of calcium in the blood
(b) Ovaries	Master gland secreting at least nine hormones, including growth hormone and TSH
(c) Pineal gland	Produces hormones involved in the regulation of metabolic rate
(d) Parathyroid glands	Secretes melatonin to regulate sleep patterns and cycles of reproductive hormones
(e) Thyroid	Produce estrogen and progesterone in response to hormones from the pituitary

3. Review the three types of stimuli for hormone release and describe a specific example of each:

(a) Hormonal stimulus: _____

(b) Humoral stimulus: _____

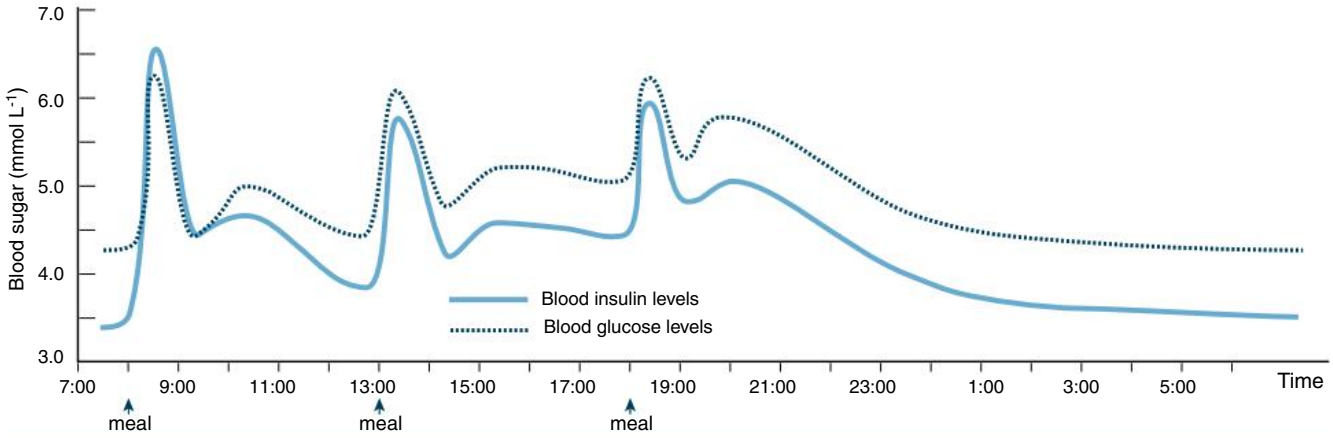
(c) Neural stimulus: _____

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53 Control of Blood Glucose

Key Idea: The endocrine part of the pancreas (the α and β cells of the islets of Langerhans) produces two hormones, glucagon and insulin, which maintain blood glucose homeostasis through negative feedback.

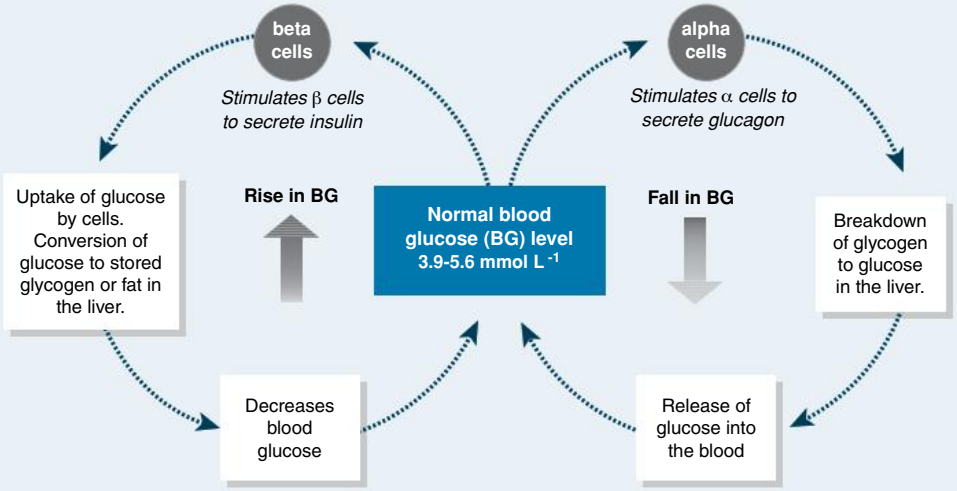
Insulin promotes a decrease in blood glucose by promoting cellular uptake of glucose and synthesising glycogen. Glucagon promotes an increase in blood glucose through the breakdown of glycogen and the synthesis of glucose from amino acids. Negative feedback stops hormone secretion when normal blood glucose levels are restored. Blood glucose homeostasis allows energy to be available to cells as needed. Extra energy is stored as glycogen or fat. These storage molecules are converted to glucose when energy is needed. The liver has a central role in these carbohydrate conversions. One of the consequences of a disruption to the insulin-glucagon system is the disease **diabetes mellitus**.





RA

In type 1 diabetes mellitus, the β cells of the pancreas are destroyed and insulin must be delivered to the bloodstream by injection. Type 2 diabetics produce insulin, but their cells do not respond to it.



1. (a) Identify the stimulus for the release of insulin: _____

   (b) Identify the stimulus for the release of glucagon: _____

   (c) Explain how glucagon brings about an increase in blood glucose level: _____

   _____

   (d) Explain how insulin brings about a decrease in blood glucose level: _____

   _____

2. Explain the pattern of fluctuations in blood glucose and blood insulin levels in the graph above:

   _____

   _____

   _____

   _____

3. The stimulus for the production and release of insulin and glucagon is: hormonal / humoral / neural (circle one):

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Type 1 Diabetes mellitus (insulin dependent)

Deficient Insulin Production

Age at onset: Early in life; often in childhood (type 1 diabetes mellitus is often called juvenile onset diabetes).

Cause: Absolute deficiency of insulin due to lack of insulin production (pancreatic β cells are destroyed in an autoimmune reaction). There is a genetic component but usually a childhood viral infection triggers the development of the disease. Mumps, coxsackie, and rubella are implicated.

Treatment: Blood glucose is monitored regularly and insulin injections combined with dietary management have been used to keep blood sugar levels stable. New therapies involving transplants of insulin-producing islet cells have been used with varying degrees of success. In the future, the option of gene therapy, where the gene for insulin production is inserted into the patient's cells, may be possible.



Type 2 Diabetes mellitus (insulin resistant)

Deficient Insulin Response

Age at onset: Historically, type 2 diabetes mellitus has usually occurred in adults over the age of 40, but its incidence is increasing in younger adults and obese children.

Cause: Type 2 diabetes may have a genetic component (in susceptibility), but it occurs most commonly as a result of lifestyle factors. Obesity (BMI > 27), a sedentary lifestyle, hypertension, high blood lipids, and a poor diet all contribute to make a person more susceptible to developing type 2 diabetes. Ethnicity may also be a contributing factor.

Treatment: Increasing physical activity, losing weight (especially abdominal fat), and improving diet may be sufficient to control type 2 diabetes. The use of prescribed anti-diabetic drugs and insulin therapy (injections) may be required if lifestyle changes are insufficient on their own.

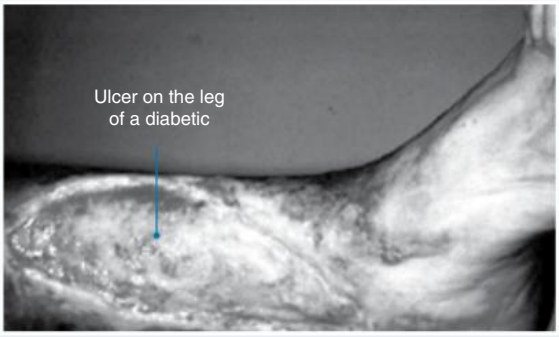


The Effects of Diabetes Mellitus

Glucose is used by cells for energy. Low insulin or insulin resistance results in low glucose within the cell. This forces cells to metabolise fats for energy. Fat metabolism produces ketones as a by-product, which can build up in the blood and cause **ketoacidosis**, with lowers the pH of the blood and can result in death.

Fats moving through the blood to places of metabolism can stick to or irritate the walls of blood vessels and cause cardiovascular diseases, such as atherosclerosis.

High blood glucose irritates and damages blood vessels and nerves. Results include numbness or tingling, loss of vision as retinal cells are damaged, gangrene and failure or wounds to heal.



4. Explain briefly why diabetes mellitus results in high blood sugar levels (hyperglycaemia): _____

5. Discuss the differences between type 1 and type 2 diabetes, including causes and treatments: _____

6. Explain why the increase in type 2 diabetes is considered epidemic in the developed world: _____

7. Describe some effects of high glucose levels in the blood: _____

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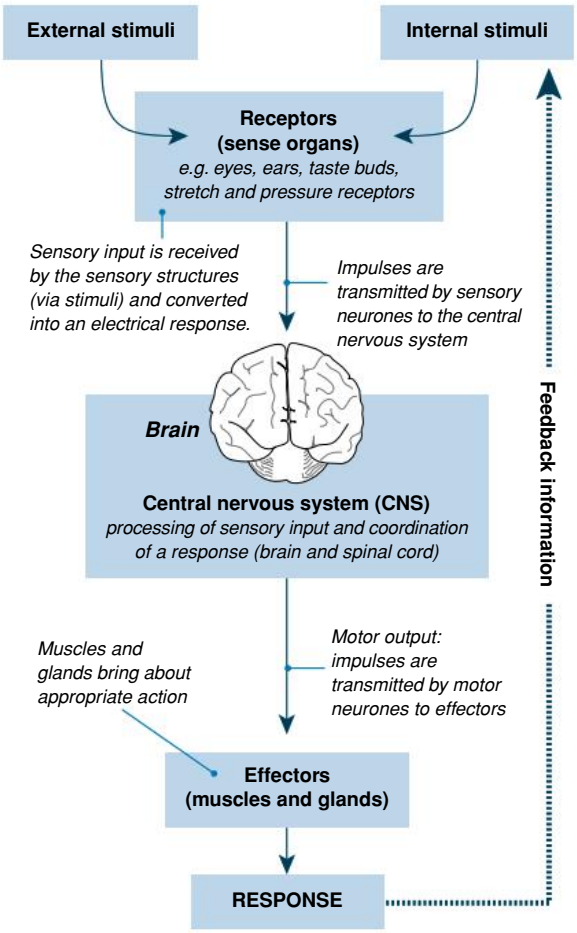
54 Nervous Regulatory Systems

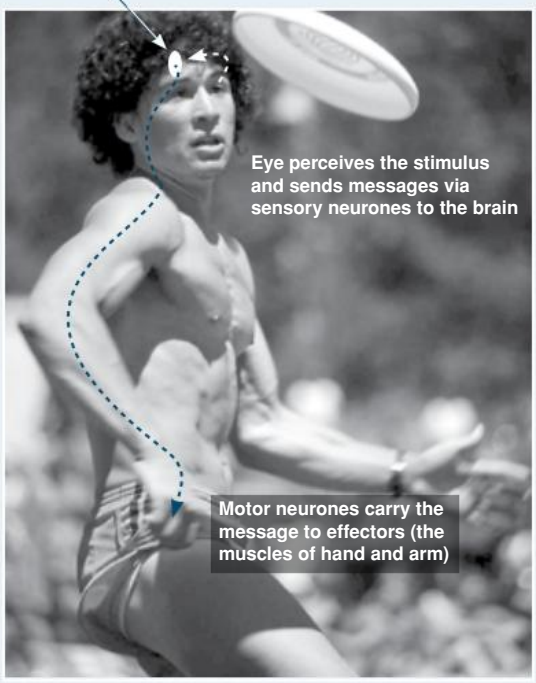
Key Idea: The nervous and endocrine systems work together to maintain homeostasis. The nervous system has three basic components: a sensory receptor, central nervous system, and effectors to bring about the response.

In mammals, the **nervous** and **endocrine** (hormonal) systems work together to maintain homeostasis. The nervous system contains cells called neurones (nerve cells) which are specialised to transmit information in the form of electrochemical impulses (action potentials). The nervous system is a signalling network with branches carrying information directly to and from specific target tissues. Impulses can be transmitted over considerable distances and the response is very precise and rapid. The basic plan of the nervous system in a human is described below, left.

Coordination by the Nervous System

The vertebrate nervous system consists of the central nervous system (brain and spinal cord), and the nerves and receptors outside it (peripheral nervous system). Sensory input to receptors comes via stimuli. Information about the effect of a response is provided by feedback mechanisms so that the system can be readjusted. The basic organisation of the nervous system can be simplified into a few key components: the sensory receptors, a central nervous system processing point, and the effectors which bring about the response (below):





In the example above, the approach of the frisbee is perceived by the eye. The motor cortex of the brain integrates the sensory message. Coordination of hand and body orientation is brought about through motor neurones to the muscles.

Comparison of nervous and hormonal control

	Nervous control	Hormonal control
Communication	*Impulses across synapses*	*Hormones in the blood*
Speed	*Very rapid (within a few milliseconds)*	*Relatively slow (over minutes, hours, or longer)*
Duration	*Short term and reversible*	*Longer lasting effects*
Target pathway	*Specific (through nerves) to specific cells*	*Hormones broadcast to target cells everywhere*
Action	*Causes glands to secrete or muscles to contract*	*Causes changes in metabolic activity*

1. Identify the three basic components of a nervous system and describe their role in maintaining homeostasis:

   (a) _____

   (b) _____

   (c) _____

2. Describe two differences between nervous control and endocrine (hormonal) control of body systems:

   (a) _____

   (b) _____

KNOW WEB 54 LINK 51 LINK 56

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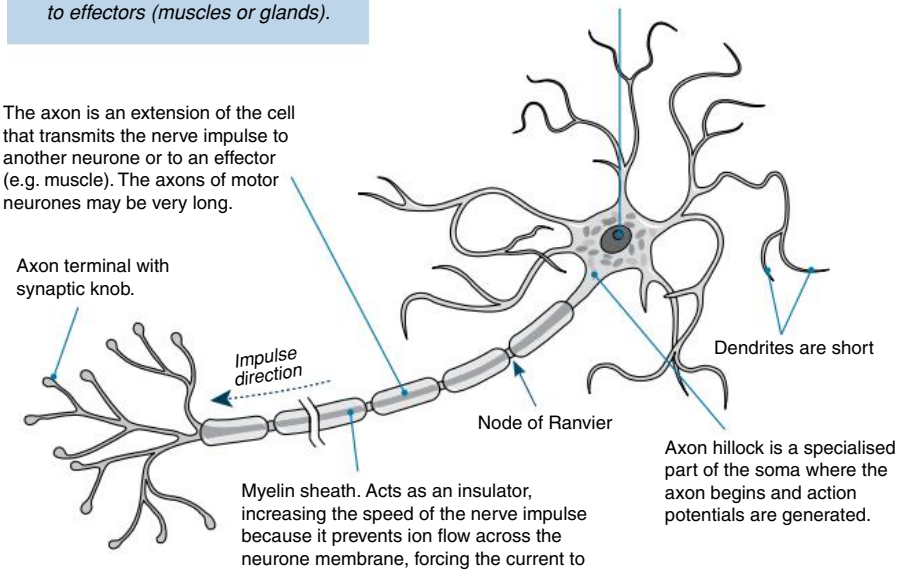
55 Neurones and Neurotransmitters

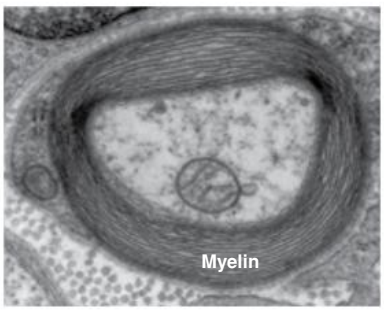
Key Idea: Neurones are electrically excitable cells that are specialised to process and transmit information via electrical and chemical signals. Neurotransmitters are chemicals that allow the transmission of signals between neurones.

Neurones are cells specialised to transmit information in the form of electrochemical signals from receptors (in the central nervous system) to effectors. Neurones consist of a cell body (soma) and long processes (dendrites and axons). Messages are transmitted between neurones by signalling molecules called neurotransmitters.

Motor (efferent) neurone

Transmits impulses from the CNS to effectors (muscles or glands).





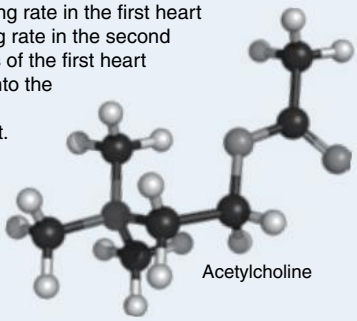
Roadnottaken

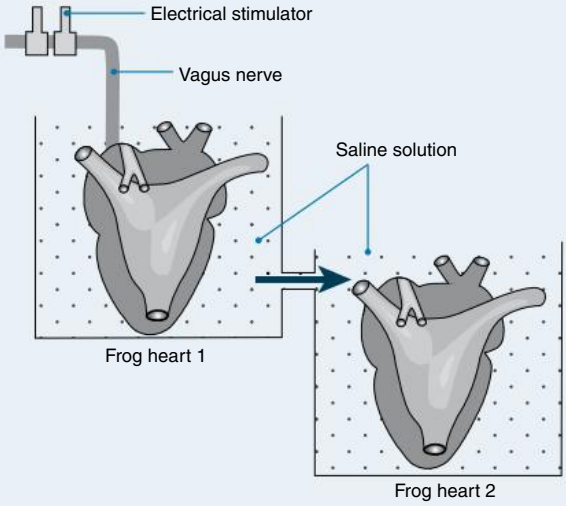
TEM cross section through a myelinated axon

Where conduction speed is important, the axons of neurones are sheathed within a lipid and protein rich substance called **myelin**. Myelin is produced by oligodendrocytes in the central nervous system (CNS) and by Schwann cells in the peripheral nervous system (PNS). At intervals along the axons of myelinated neurones, there are gaps between neighbouring Schwann cells and their sheaths. These are called **nodes of Ranvier**.

Neurotransmitters Carry Signals Between Neurones

Chemical signalling between neurones was first demonstrated in 1921 by Otto Loewi. In his experiment, the still beating hearts of two frogs were placed in connected flasks filled with saline solution. The vagus nerve (parasympathetic) of the first heart was still attached and was stimulated by electricity to reduce its rate of beating. After a delay, the rate of beating in the second heart also slowed. Increasing the beating rate in the first heart caused an increase in the beating rate in the second heart, showing electrical stimulus of the first heart caused it to release a chemical into the saline solution that then affected the heartbeat of the second heart. The chemical was found to be **acetylcholine**.





1. What is the function of a neurone? _____

_____

2. Describe the purpose of a neurotransmitter: _____

_____

3 (a) Explain why stimulating the first frog heart with electricity caused it to change its beating rate: _____

_____

_____

(b) Explain why the second heart in the experiment reduced its beating rate after a delay: _____

_____

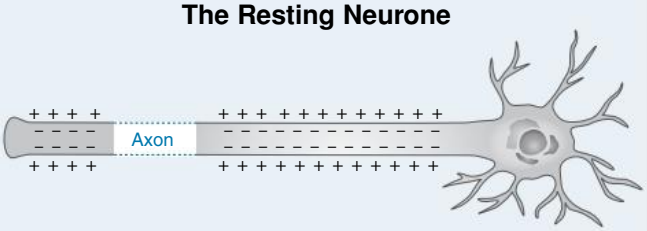
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56 Transmission of Nerve Impulses

Key idea: A nerve impulse involves the movement of an action potential along a neurone as a series of electrical depolarisation events in response to a stimulus.

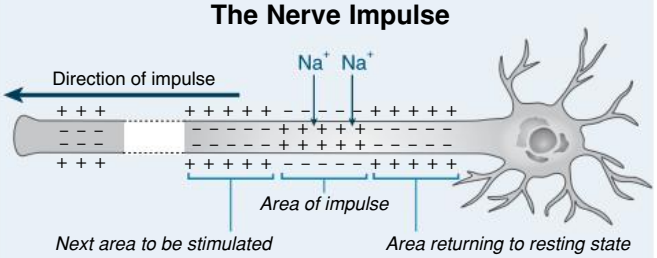
The plasma membranes of cells, including neurones, contain **sodium-potassium ion pumps** which actively pump sodium ions (Na^+) out of the cell and potassium ions (K^+) into the cell. The action of these ion pumps in neurones creates a separation of charge (a potential difference or voltage) either side of the membrane and makes the cells **electrically excitable**. It is this property that enables neurones to transmit electrical impulses. The **resting state** of a neurone, with a net negative charge inside, is maintained by the sodium-potassium pumps, which actively move two K^+ into the neurone for every three Na^+ moved out (below left). When a nerve is stimulated, a brief increase in membrane permeability to Na^+ temporarily reverses the membrane polarity (a **depolarisation**). After the nerve impulse passes, the sodium-potassium pump restores the resting potential.

The Resting Neurone

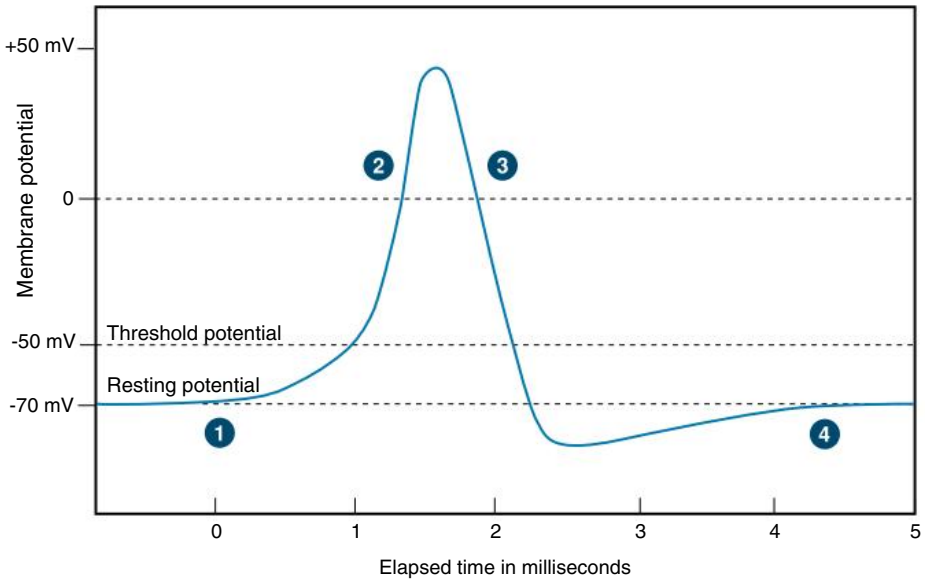


When a neurone is not transmitting an impulse, the inside of the cell is negatively charged relative to the outside and the cell is said to be electrically polarized. The potential difference (voltage) across the membrane is called the **resting potential**. For most nerve cells this is about -70 mV. Nerve transmission is possible because this membrane potential exists.

The Nerve Impulse



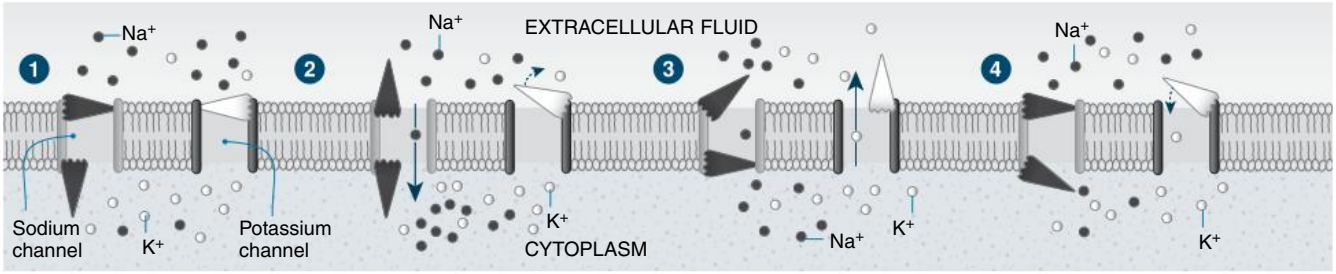
When a neurone is stimulated, the distribution of charges on each side of the membrane briefly reverses. This process of **depolarisation** causes a burst of electrical activity to pass along the axon of the neurone as an **action potential**. As the charge reversal reaches one region, local currents depolarize the next region and the impulse spreads along the axon.



The depolarisation in an axon can be shown as a change in membrane potential (in millivolts). A stimulus must be strong enough to reach the **threshold potential** before an action potential is generated. This is the voltage at which the depolarisation of the membrane becomes unstoppable.

The action potential is **all or nothing** in its generation and because of this, impulses (once generated) always reach threshold and move along the axon without attenuation. The resting potential is restored by the movement of potassium ions (K^+) out of the cell. During this **refractory period**, the nerve cannot respond, so nerve impulses are discrete.

Voltage-Gated Ion Channels and the Course of an Action Potential



Resting state:

Voltage activated Na^+ and K^+ channels are closed.

Depolarisation:

Voltage activated Na^+ channels open and there is a rapid influx of Na^+ ions. The interior of the neurone becomes positive relative to the outside.

Repolarisation:

Voltage activated Na^+ channels close and the K^+ channels open; K^+ moves out of the cell, restoring the negative charge to the cell interior.

Returning to resting state:

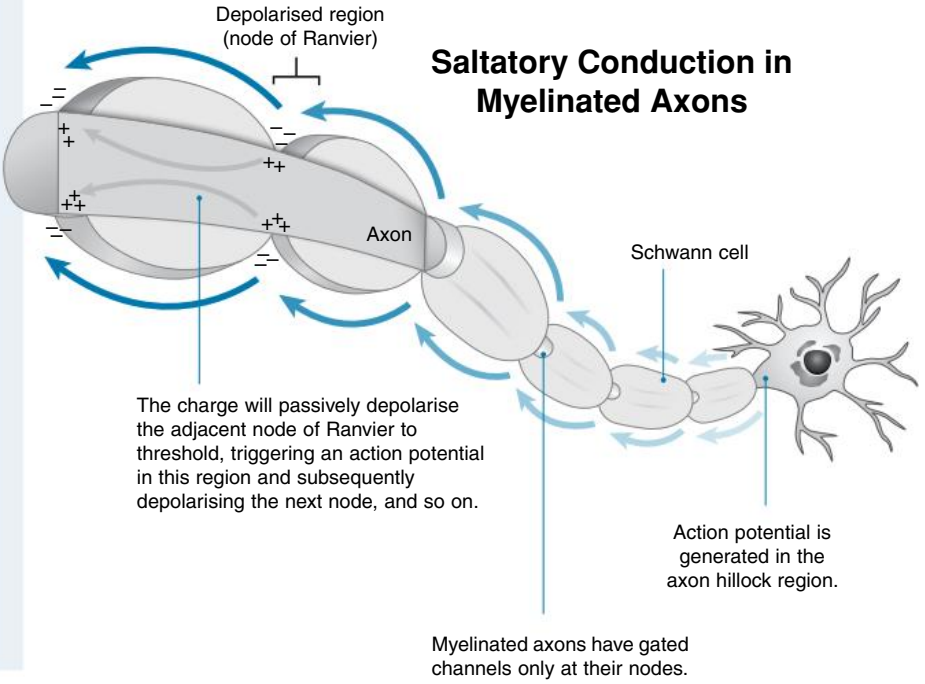
Voltage activated Na^+ and K^+ channels close to return the neurone to the resting state.

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Axon myelination is a feature of vertebrate nervous systems and it enables them to achieve very rapid speeds of nerve conduction. Myelinated neurones conduct impulses by **saltatory conduction**, a term that describes how the impulse jumps along the fibre. In a myelinated neurone, action potentials are generated only at the nodes, which is where the voltage gated channels occur. The axon is insulated so the action potential at one node is sufficient to trigger an action potential in the next node and the impulse jumps along the fibre. This differs from impulse transmission in a non-myelinated neurone in which voltage-gated channels occur along the entire length of the axon.

As well as increasing the speed of conduction, the myelin sheath reduces energy expenditure because the area over which depolarisation occurs is less (and therefore the number of sodium and potassium ions that need to be pumped to restore the resting potential is fewer).

Saltatory Conduction in Myelinated Axons



1. What is an action potential? _____

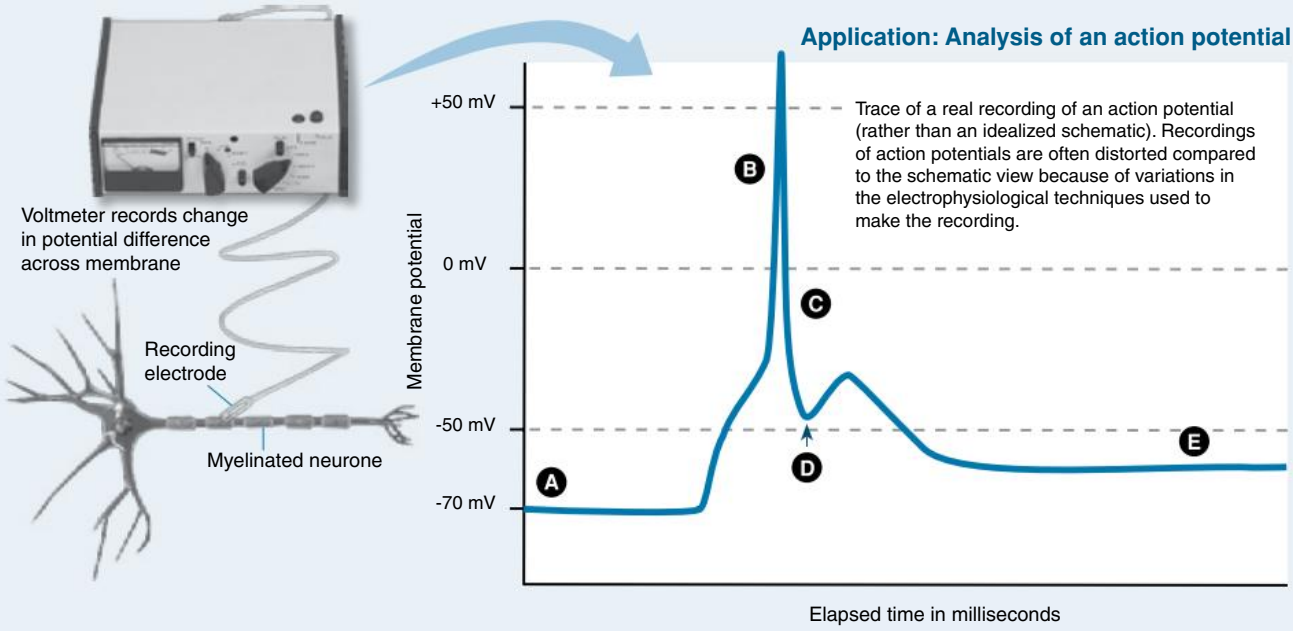
_____

2. (a) What occurs during saltatory conduction? _____

_____

(b) What influence does this have on conduction speed? _____

Application: Analysis of an action potential



3. The graph above shows a recording of the changes in membrane potential in an axon during transmission of an action potential. Match each stage (A-E) to the correct summary provided below.

- ☐ Membrane depolarisation (due to rapid Na^+ entry across the axon membrane.
- ☐ Hyperpolarisation (an overshoot caused by the delay in closing of the K^+ channels.
- ☐ Return to resting potential after the stimulus has passed.
- ☐ Repolarisation as the Na^+ channels close and slower K^+ channels begin to open.
- ☐ The membrane's resting potential.

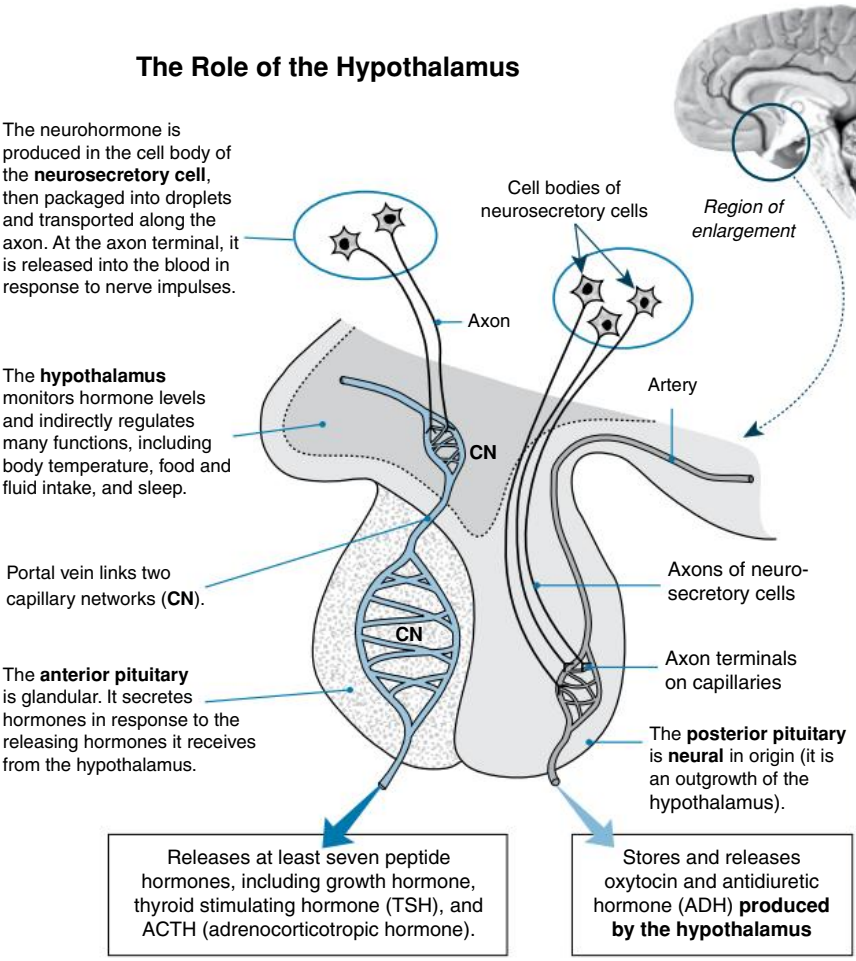
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57 Neurohormones

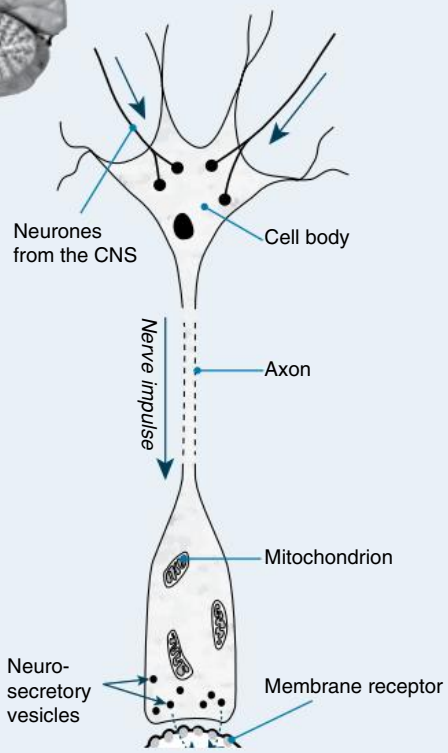
Key Idea: Neurohormones are hormones produced and released by specialised neurosecretory cells, which function as both nerve cells and endocrine cells.

The **hypothalamus** synthesises and secretes several neurohormones and links the nervous system to the endocrine system via the pituitary, with which it has a close structural and functional relationship. The **posterior pituitary** is **neural** in origin and is essentially an extension of the hypothalamus. Its neurosecretory cells release oxytocin and ADH directly into the blood in response to nerve impulses. The **anterior pituitary** is connected to the hypothalamus by blood vessels and receives neurohormones from the hypothalamus via a capillary network. These **hypothalamic releasing hormones** regulate the secretion of the anterior pituitary's hormones.

The Role of the Hypothalamus



Neurosecretory Cells



1. (a) Explain how the anterior and posterior pituitary differ with respect to their relationship to the hypothalamus:

   _____

   _____

   (b) Explain how these differences relate to the nature of the hormonal secretions for each region: _____

   _____

   _____

2. Describe the role of the neurohormones released by the hypothalamus: _____

   _____

3. Suggest why the thyroid gland will atrophy (waste away) if the pituitary gland ceases to function: _____

   _____

   _____

4. Although the anterior pituitary is often called the master gland, the hypothalamus could also claim that title. Explain:

   _____

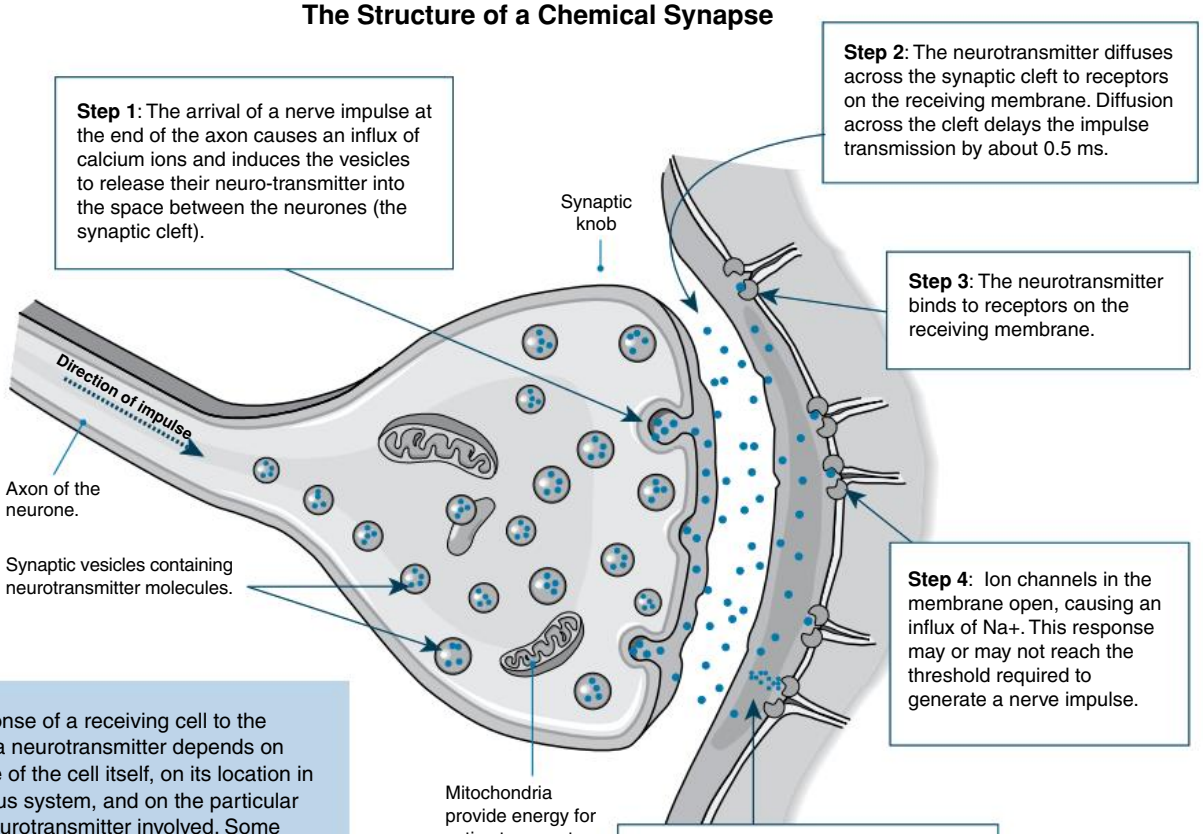
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58 Chemical Synapses

Key Idea: Chemical synapses are junctions between neurones, or between neurones and receptor or effector cells. Action potentials are transmitted across junctions called **synapses**. Synapses can occur between two neurones, or between a neurone and an effector cell (e.g. muscle or gland). The axon terminal is a swollen knob, and a small gap (synaptic cleft) separates it from the receiving neurone. The synaptic knobs are filled with tiny packets of chemicals called **neurotransmitters**. The neurotransmitter diffuses across the gap, where it interacts with the receiving (post-synaptic) membrane and causes an electrical response. In the example below, the neurotransmitter causes a membrane depolarisation and the generation of an action potential. Some neurotransmitters have the opposite effect and cause inhibition (e.g. slowing heart rate). Chemical synapses are the most widespread type of synapse in nervous systems.

The Structure of a Chemical Synapse



The response of a receiving cell to the arrival of a neurotransmitter depends on the nature of the cell itself, on its location in the nervous system, and on the particular type of neurotransmitter involved. Some examples of neurotransmitters include: acetylcholine and noradrenaline (in the peripheral nervous system), and GABA and serotonin, (in the brain).

1. What is a **synapse**? _____

2. What causes the release of neurotransmitter into the synaptic cleft? _____

_____

3. Why is there a brief delay in transmitting an impulse across the synapse? _____

_____

4. (a) How is the neurotransmitter deactivated? _____

(b) Why is it important for the neurotransmitter substance to be deactivated soon after its release? _____

_____

5. Consult a reference source to identify one function of acetylcholine in the nervous system: _____

_____

6. Suggest one factor that might influence the strength of the response in the receiving cell: _____

_____

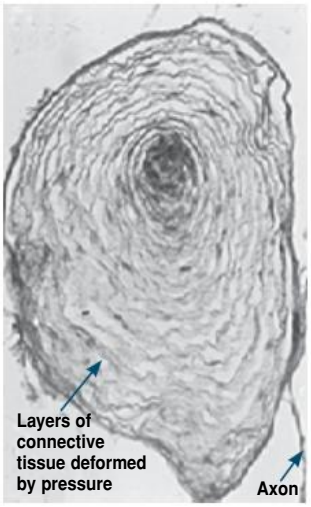
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59 Signal Transduction in Sense Organs

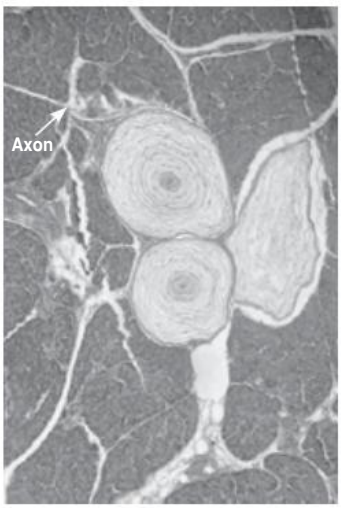
Key Idea: Sensory receptors convert stimulus energy into an electrochemical signal, which can be interpreted. Sensory receptors are specialised to detect stimuli and respond by producing an electrical discharge. In this way they act as **biological transducers**, converting the energy from a stimulus into an electrochemical signal. Stimulation of a sensory receptor cell results in an electrical impulse with specific properties. The frequency of impulses produced by the receptor cell encodes information about the strength of the stimulus. A stronger stimulus produces more frequent impulses. Sensory receptors also show **sensory adaptation** and will cease responding to a stimulus of the same intensity. Sensory receptors are classified according to the stimuli to which they respond (e.g. photoreceptors respond to light). The response of a simple **mechanoreceptor**, the Pacinian corpuscle, to a stimulus (pressure) is described below.

The Pacinian Corpuscle

Pacinian corpuscles are pressure receptors occurring in deep subcutaneous tissues all over the body. They are relatively large and simple in structure, consisting of a sensory nerve ending (dendrite) surrounded by a capsule of layered connective tissue. Pressure deforms the capsule, stretching the nerve ending and leading to a localised depolarisation. Once a **threshold** value is reached, an **action potentia**l propagates along the axon.

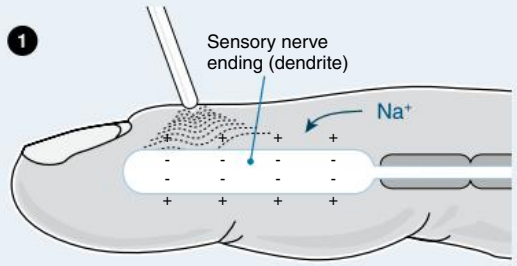


RCN

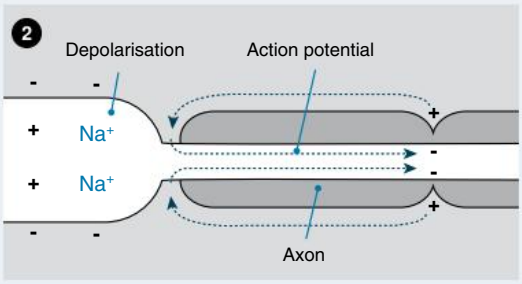


D. Fankhauser, University of Cincinnati, Clermont College

Pacinian corpuscle (above, left), illustrating the distinctive layers of connective tissue. The photograph on the right shows corpuscles grouped together in the pancreas. Pacinian corpuscles are rapidly adapting receptors; they fire at the beginning and end of a stimulus, but do not respond to unchanging pressure.



Deforming the corpuscle leads to an increase in the permeability of the nerve to sodium. Na^+ diffuses into the nerve ending creating a localised depolarisation. This depolarisation is called a **generator potential**.



A volley of **action potentials** is triggered once the generator potential reaches or exceeds a **threshold value**. These action potentials are conducted along the sensory axon. A strong stimulus results in a high frequency of impulses.

1. Why are sensory receptors termed **biological transducers**? _____

2. What is the significance of linking the magnitude of a sensory response to stimulus intensity? _____

3. What is the physiological importance of sensory adaptation? _____

4. (a) Describe the properties of a generator potential: _____

   (b) Why must the stimulus strength reach a threshold value before the sensory receptor will respond?

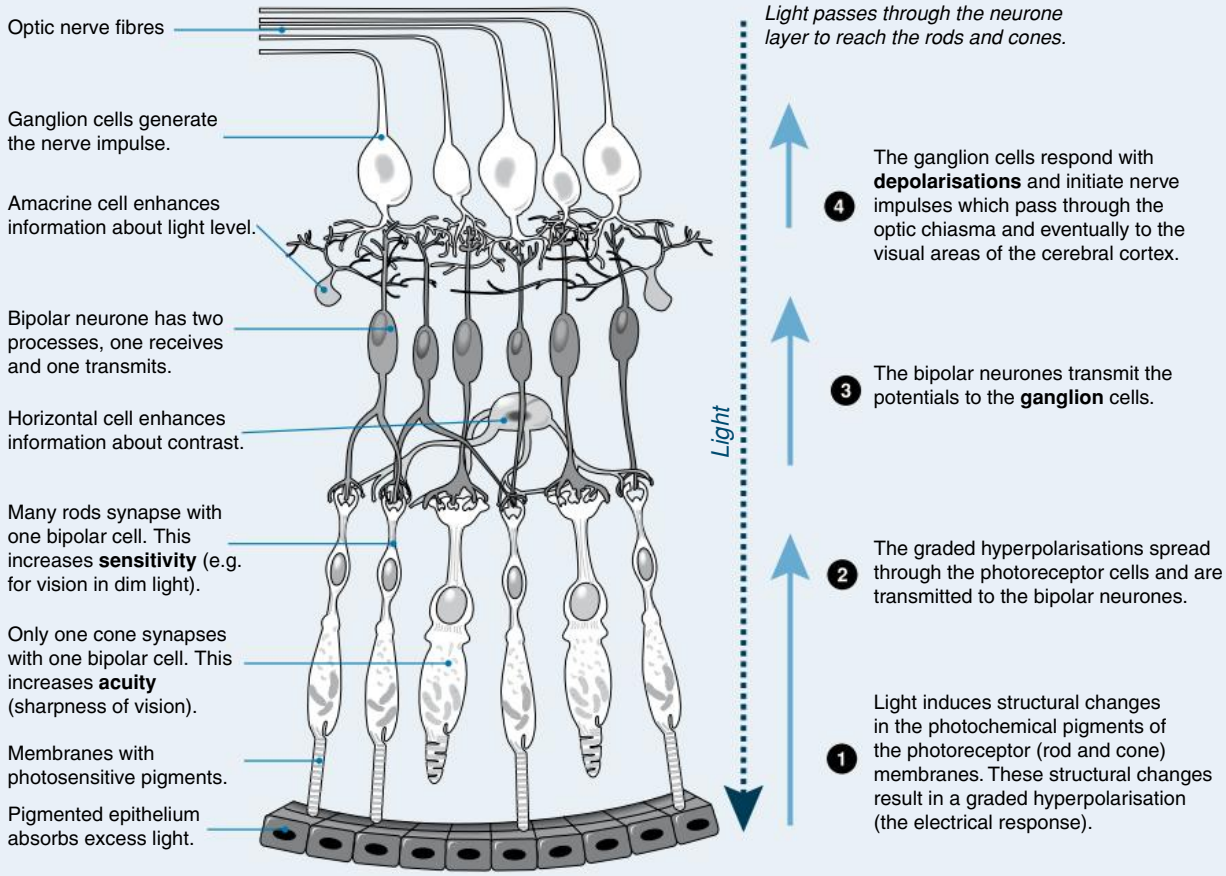
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60 The Physiology of Vision

Key Idea: The eye is a sensory organ that converts light into nerve impulses resulting in the formation of a visual image.

Sense organs are collections of sensory receptors with their accessory structures. An example of such an organ is the eye. In the mammalian eye, vision is achieved by focussing the light through a lens to form an image on the back of the eye (the **retina**). When light reaches the retina, it is absorbed by the photosensitive pigments associated with the membranes of the **photoreceptor cells** (the rods and cones). The pigment molecules are altered by the absorption of light, producing nerve impulses. The electrical signals are transmitted from the eye via the **optic nerve**, along the visual pathway to the visual cortex of the brain, where the information is interpreted. The retina is not uniform. The **central fovea** is an area where there is a high density of cones and virtually no rods. It is the region of highest acuity.

Structure and Function of the Retina



1. Describe the role of each of the following in human vision:

(a) Retina: _____

_____

_____

_____

(b) Optic nerve: _____

_____

_____

_____

(c) Central fovea: _____

_____

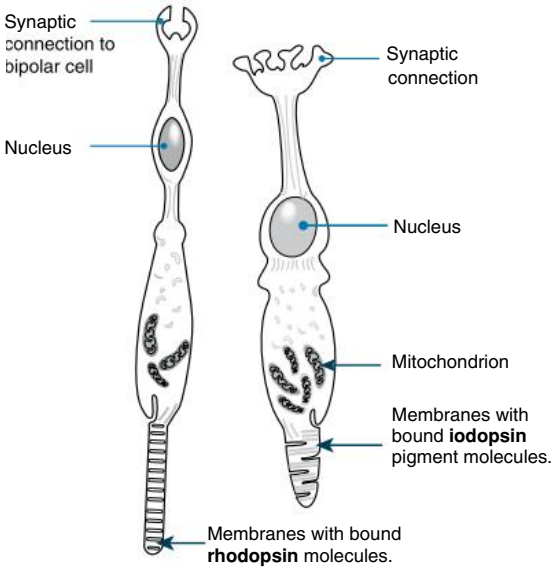
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Photograph through the eye of a normal retina. The blind spot, where the where ganglion cell axons exit the eye to form the optic nerve, is seen as the bright area to the left of the image. The central fovea, where cone density is highest, is in the darker region at the centre of the image. Note the rich blood supply.

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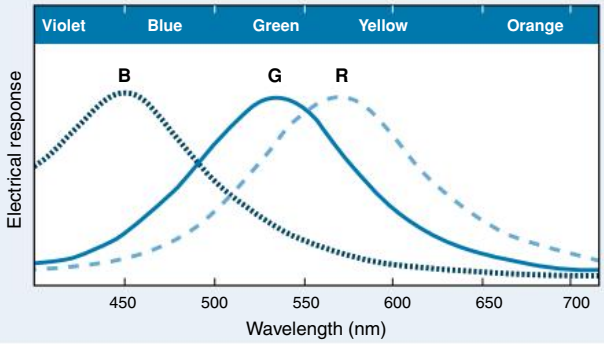


Structure of rod (left) and cone (right) photoreceptor cells

The Basis of Trichromatic Vision

There are three classes of cones, each with a maximal response in either short (blue), intermediate (green) or long (yellow-green) wavelength light (below). The yellow-green cone is also sensitive to the red part of the spectrum and is often called the red cone. The differential responses of the cones to light of different wavelengths provides the basis of trichromatic colour vision.

Cone response to light wavelengths



2. Complete the table below, comparing the features of rod and cone cells:

Feature	Rod cells	Cone cells
Visual pigment(s):		
Visual acuity:		
Overall function:		

3. Identify the three major types of neurone making up the retina and describe their basic function:

   (a) _____

   (b) _____

   (c) _____

4. Identify two types of accessory neurones in the retina and describe their basic function:

   (a) _____

   (b) _____

5. Account for the differences in acuity (sharpness of vision) and sensitivity (to light level) between rod and cone cells:

   _____

   _____

6. (a) What is meant by the term **photochemical pigment** (photopigment)? _____

   _____

   _____

   (b) Identify two photopigments and their location: _____

   _____

7. In your own words, explain how light is able to produce a nerve impulse in the ganglion cells: _____

   _____

   _____

   _____

   _____

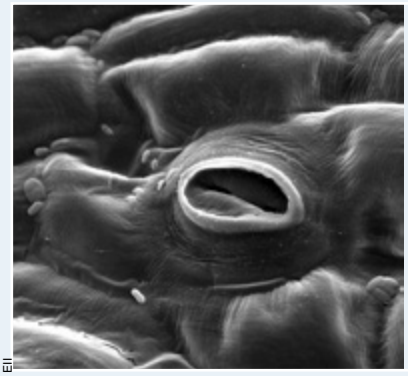
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61 Auxins, Gibberellins, and ABA

Key Idea: Phytohormones, such as auxin, gibberellin, and ABA, play important roles in plant responses to stimuli.

Phytohormones (plant growth regulators) have a wide range of roles in the growth and developmental responses of vascular plants. **Auxin** (indole-acetic acid or IAA) is a naturally occurring phytohormone with a role in suppressing the growth of lateral buds. This inhibitory influence of a shoot tip or apical bud on the lateral buds is called apical dominance. **Gibberellins** are involved in stem and leaf elongation, as well as breaking dormancy in seeds. Specifically, they stimulate cell division and cell elongation. **Abscisic** acid or (ABA) was originally thought to be involved in accelerating abscission in leaves and fruit. Although it now seems that ABA is a growth inhibitor with a variety of roles.

Plant Responses and the Role of Auxins, Gibberellins, and Abscisic Acid



ABA stimulates the closing of stomata in most plant species. Its synthesis is stimulated by water deficiency (water stress). ABA also promotes seed dormancy. It is concentrated in senescent leaves, but it is probably not involved in leaf abscission except in a few species.



Gibberellins are responsible for breaking dormancy in seeds and promote the growth of the embryo and emergence of the seedling. Gibberellins are used to hasten seed germination and ensure germination uniformity in the production of barley malt in brewing.



Differential auxin transport, and therefore differential growth, is responsible for phototropism. Auxins also produce apical dominance in shoots. Auxin is produced in the shoot tip and diffuses down the stem to inhibit the development of the lateral buds.



ABA is produced in ripe fruit and induces fruit fall. The effects of ABA are generally opposite to those of cytokinins.



Gibberellins cause stem and leaf elongation by stimulating cell division and cell elongation. They are responsible for bolting in brassicas.



Auxin is required for fruit growth. The maturing seed releases auxin, inducing the surrounding flower parts to develop into fruit.

1. What is the role of **gibberellins** in stem elongation and in the germination of grasses such as barley?

2. What is the role of **abscisic acid** in closure of stomata?

3. Why is abscisic acid often referred to as a growth inhibitor?

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LINK 62 KNOW

62 Plant Hormones as Signal Molecules

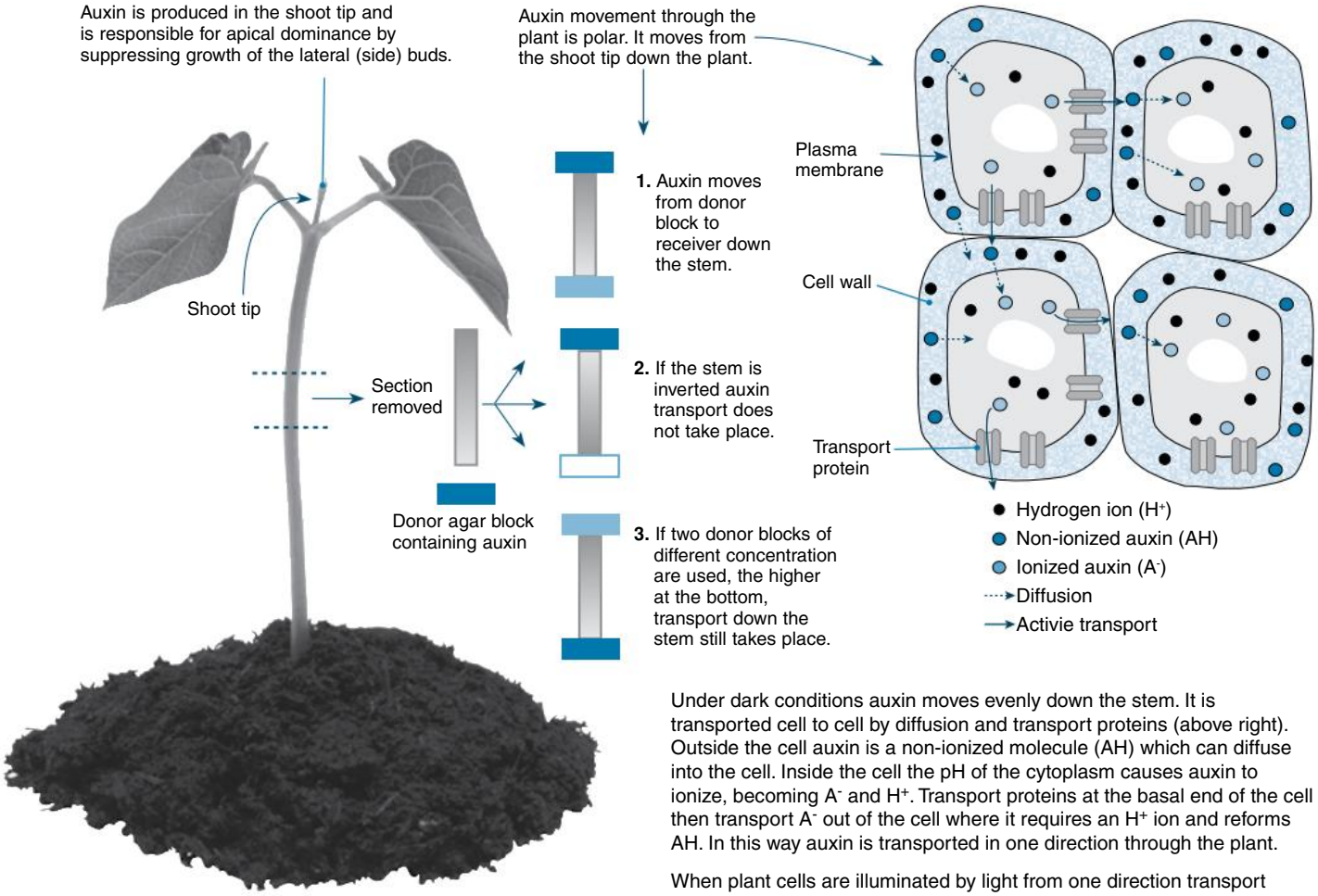
Key Idea: Auxin is a plant hormone involved in the differential growth responses of plants to environmental stimuli.

Auxins have a central role in a wide range of growth and developmental responses in vascular plants. Indole-acetic acid (IAA) is the most potent native auxin in intact plants. The response of a plant tissue to IAA depends on the tissue itself, the concentration of the hormone, the timing of its release, and the presence of other phytohormones. Gradients in auxin concentration during growth prompt differential responses in specific tissues and contribute to directional growth.

Light is an important growth requirement for all plants. Most plants show an adaptive response of growing towards the light. This growth response is called phototropism. A **tropism** is a plant growth response to external stimuli in which the stimulus direction determines the direction of the growth response.



Tropisms are identified according to the stimulus involved, e.g. photo- (light) and gravi- (gravity), and may be positive or negative depending on whether the plant moves towards or away from the stimulus respectively. The bending of the plants shown on the right is a phototropism in response to light shining from the left and caused by the plant hormone **auxin**. Auxin causes the elongation of cells on the shaded side of the stem, causing it to bend.



Under dark conditions auxin moves evenly down the stem. It is transported cell to cell by diffusion and transport proteins (above right). Outside the cell auxin is a non-ionized molecule (AH) which can diffuse into the cell. Inside the cell the pH of the cytoplasm causes auxin to ionize, becoming A^- and H^+. Transport proteins at the basal end of the cell then transport A^- out of the cell where it requires an H^+ ion and reforms AH. In this way auxin is transported in one direction through the plant.

When plant cells are illuminated by light from one direction transport proteins in the plasma membrane on the shaded side of the cell are activated and auxin is transported to the shaded side of the plant.

1. What is the term given to the tropism being displayed in the photo (top right)? _____
2. Describe one piece of evidence that demonstrates the transport of auxin is polar: _____

_____

_____

_____

3. What is the effect of auxin on cell growth? _____

_____

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63 Signals for Daylength in Plants

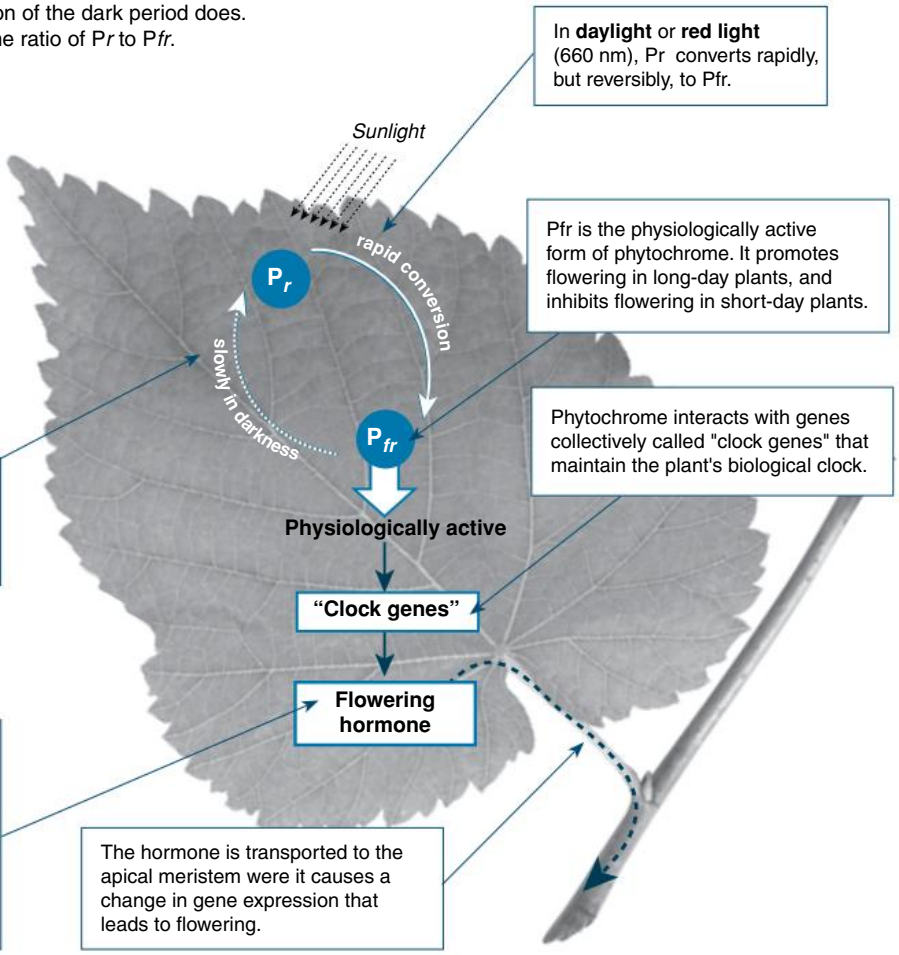
Key Idea: Photoperiodism is the response of a plant to the relative lengths of light and dark. It is controlled by the pigment phytochrome, which occurs in two forms P*r* and P*fr*. Flowering is a photoperiodic activity that is dependent on the species' response to light. It is controlled through the action of a pigment called **phytochrome**. Phytochrome acts as a signal for some biological clocks in plants and exists in two forms, P*r* and P*fr*. It is important in initiating flowering in long-day and short-day plants, but is also involved in other light initiated responses, such as germination and shoot growth.

Long-Day and Short-Day Plants

Short-day plants flower when the photoperiod is less than a critical length of time, while long-day plants flower when the photoperiod is greater than a critical length of time. The interruption of the light period does not inhibit flowering in short-day plants, but the interruption of the dark period does. Plants can monitor the light-dark cycle using the ratio of P*r* to P*fr*.

Phytochrome

Phytochrome is a blue-green pigment that acts as a photoreceptor for detection of night and day in plants and is universal in vascular plants. It has two forms: **P*r*** (inactive) and **P*fr*** (active). P*r* is readily converted to P*fr* under natural light. P*fr* converts back to P*r* in the dark but more slowly. P*fr* predominates in daylight. The plant measures daylength (or rather night length) by the amount of phytochrome in each form.



1. (a) Identify the two forms of phytochrome and the wavelengths of light they absorb: _____

_____

(b) Identify the biologically active form of phytochrome and how it behaves in long day plants and short day plants with respect to flowering:

_____

_____

2. (a) Discuss the role of phytochrome in a plant's ability to measure daylength: _____

_____

_____

_____

(b) How does this help coordinate flower production in a plant species? _____

_____

_____

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64 The Homeostatic Role of Blood

Key Idea: Blood is a complex liquid tissue made up of cellular components suspended in plasma. Its roles in homeostasis include thermoregulation and transport of substances.

Blood is a liquid tissue central to the body's homeostasis. It transports nutrients, respiratory gases, hormones, and wastes, it has a role in thermoregulation through the distribution of heat, it defends against infection, and its ability to clot protects against blood loss. If a blood sample is taken, the cells can be separated from the plasma by centrifugation. The cells (formed elements) settle as a dense red pellet below the transparent, straw-coloured plasma. The examination of blood is useful in diagnosing disease. The cellular components of blood are normally present in particular specified ratios. A change in the morphology, type, or proportion of different blood cells can therefore be used to indicate a specific disorder or infection.

Non-Cellular Blood Components

The non-cellular blood components form the plasma. Plasma is a watery matrix of ions and proteins and makes up 50-60% of the total blood volume.

Water

The main constituent of blood and lymph.
Role: Transports dissolved substances. Provides body cells with water. Distributes heat and has a central role in thermoregulation. Regulation of water content helps to regulate blood pressure and volume.

Mineral Ions

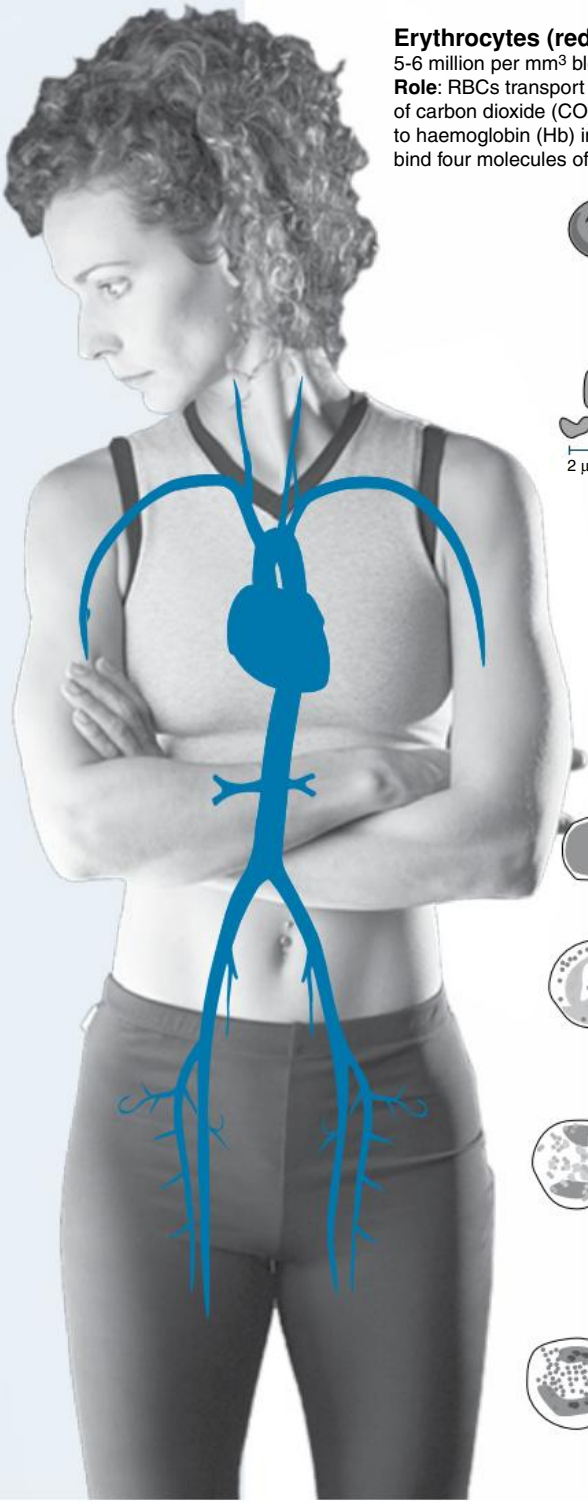
Sodium, bicarbonate, magnesium, potassium, calcium, chloride.
Role: Osmotic balance, pH buffering, and regulation of membrane permeability. They also have a variety of other functions, e.g. Ca^{2+} is involved in blood clotting.

Plasma proteins

7-9% of the plasma volume.
Serum albumin
Role: Osmotic balance and pH buffering, Ca^{2+} transport.
Fibrinogen and prothrombin
Role: Take part in blood clotting.
Immunoglobulins
Role: Antibodies involved in the immune response.
α-globulins
Role: Bind/transport hormones, lipids, fat soluble vitamins.
β-globulins
Role: Bind/transport iron, cholesterol, fat soluble vitamins.
Enzymes
Role: Take part in and regulate metabolic activities.

Substances transported by non-cellular components

Products of digestion
Examples: sugars, fatty acids, glycerol, and amino acids.
Excretory products
Example: urea
Hormones and vitamins
Examples: insulin, sex hormones, vitamins A and B12.
Importance: These substances occur at varying levels in the blood. They are transported to and from the cells dissolved in the plasma or bound to plasma proteins.

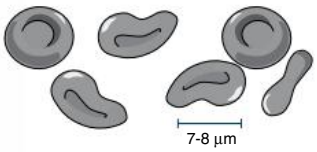


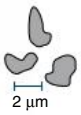
Cellular Blood Components

The cellular components of the blood (also called the formed elements) float in the plasma and make up 40-50% of the total blood volume.

Erythrocytes (red blood cells or RBCs)

5-6 million per mm^3 blood; 38-48% of total blood volume.
Role: RBCs transport oxygen (O_2) and a small amount of carbon dioxide (CO_2). The oxygen is carried bound to haemoglobin (Hb) in the cells. Each Hb molecule can bind four molecules of oxygen.





Platelets

Small, membrane-bound cell fragments derived from bone marrow cells; about 1/4 the size of RBCs.
0.25 million per mm^3 blood.
Role: To start the blood clotting process.

Leucocytes (white blood cells)

5-10 000 per mm^3 blood
2-3% of total blood volume.
Role: Involved in internal defence. There are several types of white blood cells (see below).



Lymphocytes
T and B cells.
24% of the white cell count.
Role: Antibody production and cell mediated immunity.



Neutrophils
Phagocytes.
70% of the white cell count.
Role: Engulf foreign material.



Eosinophils
Rare leucocytes; normally 1.5% of the white cell count.
Role: Mediate allergic responses such as hayfever and asthma.

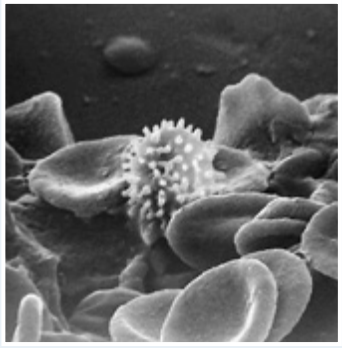


Basophils
Rare leucocytes; normally 0.5% of the white cell count.
Role: Produce heparin (an anti-clotting protein), and histamine. Involved in inflammation.

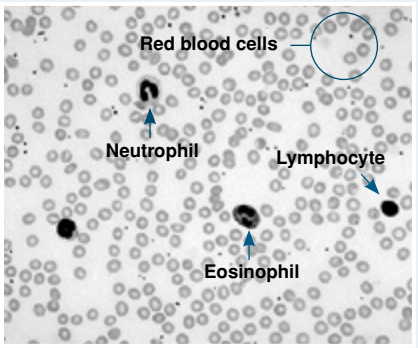
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The Examination of Blood

Different types of microscopy give different information about blood. A SEM (right) shows the detailed external morphology of the blood cells. A fixed smear of a blood sample viewed with a light microscope (far right) can be used to identify the different blood cell types present, and their ratio to each other. Determining the types and proportions of different white blood cells in blood is called a **differential white blood cell count**. Elevated counts of particular cell types indicate allergy or infection.



SEM of red blood cells and a leucocyte.



Light microscope view of a fixed blood smear.

Photos: EII

1. For each of the following homeostatic functions of blood, identify the component (or components) of the blood responsible and state how the function is carried out (the mode of action). The first one is done for you:

   (a) **Temperature regulation**. *Blood component*: Water component of the plasma

   Mode of action: Water absorbs heat and dissipates it from sites of production (e.g. organs)

   (b) **Protection against disease**. *Blood component*: _____

   Mode of action: _____

   (c) **Communication between cells, tissues, and organs**. *Blood component*: _____

   Mode of action: _____

   (d) **Oxygen transport**. *Blood component*: _____

   Mode of action: _____

   (e) **CO_2 transport**. *Blood components*: _____

   Mode of action: _____

   (f) **Buffer against pH changes**. *Blood components*: _____

   Mode of action: _____

   (g) **Nutrient supply**. *Blood component*: _____

   Mode of action: _____

   (h) **Tissue repair**. *Blood components*: _____

   Mode of action: _____

   (i) **Transport of hormones, lipids, and fat soluble vitamins**. *Blood component*: _____

   Mode of action: _____

2. Identify a feature that distinguishes red and white blood cells: _____

3. Describe two physiological advantages of red blood cell structure (lacking nucleus and mitochondria):

   (a) _____

   (b) _____

4. What might each of the following results from a differential white blood cell count suggest to you?

   (a) Elevated levels of eosinophils (above the normal range): _____

   (b) Elevated levels of neutrophils (above the normal range): _____

   (c) Elevated levels of basophils (above the normal range): _____

   (d) Elevated levels of lymphocytes (above the normal range): _____

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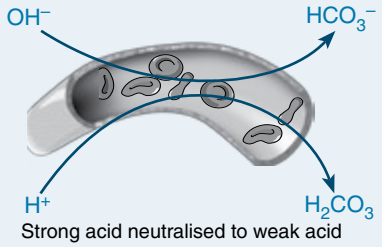
65 Acid–Base Balance

Key Idea: The narrow pH range (pH 7.35-7.45) of the body's fluids is maintained by the blood, lungs, and kidneys.

The products of metabolic activity are generally acidic and could alter pH considerably without a buffer system to counteract pH changes. The carbonic acid-bicarbonate buffer works throughout the body to maintain the pH of blood plasma close to 7.40. The body maintains the buffer by eliminating either the acid (carbonic acid) or the base (bicarbonate ions). The blood buffers, the lungs, and the kidneys represent the three defence systems against disturbances of pH homeostasis. Changes in carbonic acid concentration can be brought about within seconds via increased or decreased rate of breathing. The renal system, acts more slowly, controlling pH by excreting or retaining ions.

The Blood Buffer System

Strong base neutralised to weak base



Strong acid neutralised to weak acid

A buffer is able to resist changes to the pH of a fluid when either an acid or base is added to it. The bicarbonate ion (HCO_3^-) and its acid, carbonic acid (H_2CO_3), work in the following way:

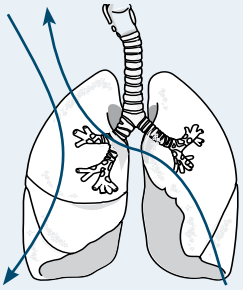
$$H^+ + HCO_3^- \rightleftharpoons H_2CO_3$$

$$H_2CO_3 \rightleftharpoons H^+ + HCO_3^-$$

If a strong acid (such as HCl) is added to the system a weak acid is formed and thus the pH falls only slightly. Note that the blood also contains proteins, which contain basic and acidic groups that may act either as H^+ acceptors or donors to help maintain blood pH.

The Respiratory System

Signal to brain $CO_2 + H_2O \rightleftharpoons H_2CO_3$



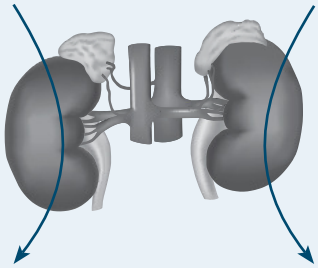
Increase in breathing rate $H_2CO_3 \rightleftharpoons H^+ + HCO_3^-$

Carbon dioxide (CO_2) in the blood, an end-product of cellular respiration, forms carbonic acid (H_2CO_3) which dissociates to form H^+ and bicarbonate (HCO_3^-). This means that as CO_2 rises in the blood so too does the H^+ concentration. **Chemoreceptors** in the brain detect the rise in H^+ ions and increase the rate of breathing to expel the CO_2. Low levels of CO_2 have the effect of depressing the respiratory system so that H^+ builds up and the pH is once again restored.

The Renal System

Rise in pH stimulates: Retain H^+

Fall in pH stimulates: Removal H^+



Equates to removal HCO_3^-

Gain HCO_3^-

Recall that a net loss of HCO_3^- effectively results in the gain of H^+. Bicarbonate is reabsorbed by the kidney tubules all the time so pH is regulated through retention or secretion of H^+. When blood pH rises, H^+ is retained by the tubule cells. When blood pH falls, H^+ is actively secreted into the kidney tubules. Urine pH normally varies from 4.5 to 8.0, reflecting the ability of the renal tubules to lose or retain ions to maintain the homeostasis of blood pH.

1. Explain why the blood must be kept at a pH between 7.35 and 7.45: _____

2. A drop in the blood pH to below 7.35 is called metabolic acidosis, even though the blood might still be at pH >7 and not strictly acidic. Describe how metabolic acidosis might arise:

3 (a) Describe how the blood buffer system maintains blood pH: _____

(b) Explain the effects of adding a base (e.g. ingestion of alkaline substances) to the system: _____

4. (a) Describe the respiratory response to excess H^+ in the blood: _____

(b) Explain where these H^+ ions come from: _____

(c) Describe how **respiratory acidosis** might arise: _____

5. Explain the role of the renal system in maintaining the pH of the blood: _____

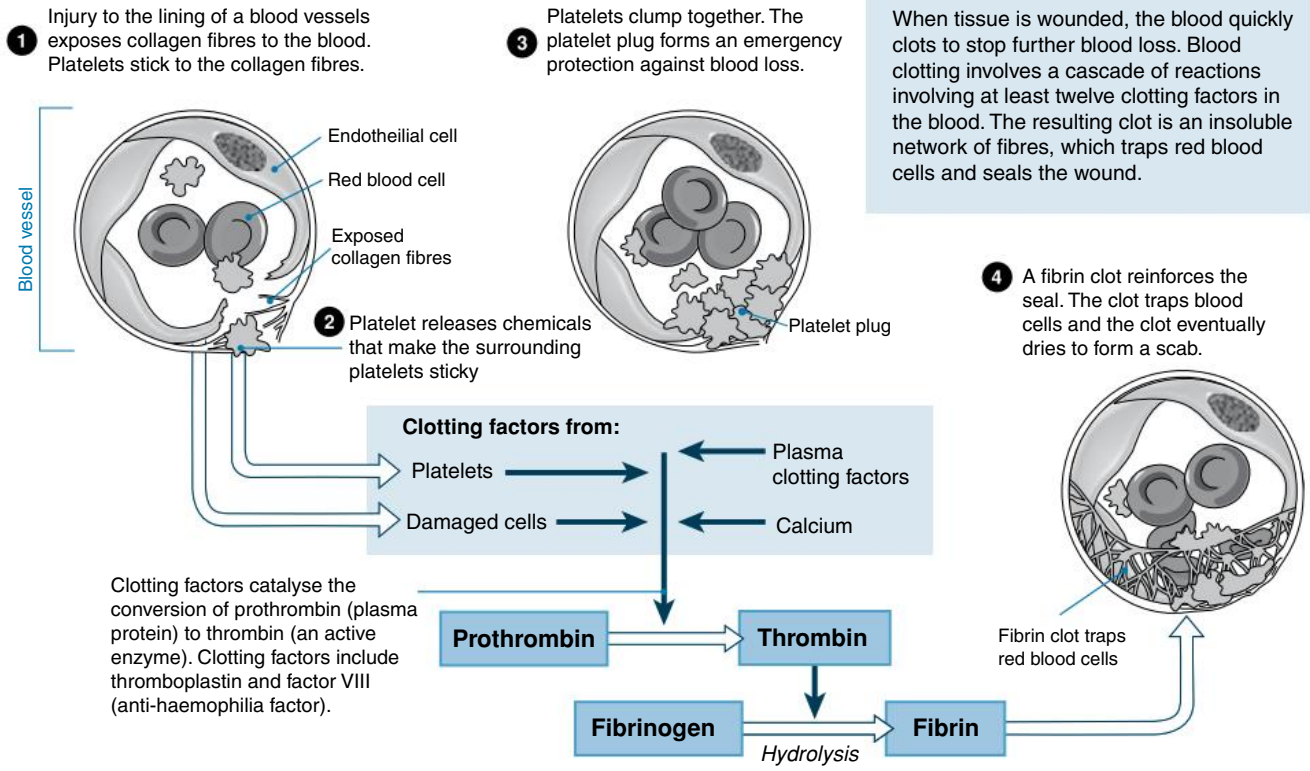
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66 Blood Clotting

Key Idea: Blood clotting restricts blood loss from a torn blood vessel, and prevents pathogens entering the wound.

Apart from its transport role, **blood** has a role in the body's defence against infection and **hemostasis** (the prevention of bleeding and maintenance of blood volume). The tearing or puncturing of a blood vessel initiates **clotting**. Clotting is a rapid process that seals off the tear, preventing blood loss and the invasion of bacteria into the site. Clotting is the result of a **positive feedback loop** initiated by the release of chemicals from injured tissue. The chemicals activate platelets, which release chemicals to activate more platelets, causing a rapid cascade that ends with the formation of the clot.

When tissue is wounded, the blood quickly clots to stop further blood loss. Blood clotting involves a cascade of reactions involving at least twelve clotting factors in the blood. The resulting clot is an insoluble network of fibres, which traps red blood cells and seals the wound.



1. Explain two roles of the blood clotting system in internal defence and haemostasis:

   (a) _____

   (b) _____

2. Explain the role of each of the following in the sequence of events leading to a blood clot:

   (a) Injury: _____

   (b) Release of chemicals from platelets: _____

   _____

   (c) Clumping of platelets at the wound site: _____

   _____

   (d) Formation of a fibrin clot: _____

   _____

3. (a) What is the role of clotting factors in the blood in formation of the clot? _____

   _____

   (b) Why are these clotting factors not normally present in the plasma? _____

   _____

4. (a) Name one inherited disease caused by the absence of a clotting factor: _____

   (b) Name the clotting factor involved: _____

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67 Thermoregulation in Mammals

Key Idea: In humans, the temperature regulation centre is in the hypothalamus. Thermoregulation relies on negative feedback mechanisms and involves several body systems.

In placental mammals the temperature regulation centre of the body is in the **hypothalamus**. It has a 'set point' temperature of 36.7°C. The hypothalamus responds directly to changes in core temperature and to nerve impulses from receptors in the skin. It then coordinates appropriate nervous and hormonal responses to counteract the changes and restore normal body temperature. Like a thermostat, the hypothalamus detects a return to normal temperature and the corrective mechanisms are switched off (negative feedback).

Counteracting Heat Loss

Heat promoting centre* in the hypothalamus monitors fall in skin or core temperature below 35.8°C and coordinates responses that generate and conserve heat. These responses are mediated primarily through the **sympathetic nerves** of the autonomic nervous system.

Thyroxine (together with adrenaline) **increases metabolic rate**.

Under conditions of *extreme* cold, adrenaline and thyroxine increase the energy releasing activity of the liver. Under normal conditions, the liver is thermally neutral.

Muscular activity (including **shivering**) produces internal heat.

Erector muscles of hairs contract to raise hairs and increase insulating layer of air. Blood flow to skin decreases (**vasoconstriction**).

Factors causing heat loss

- Wind chill factor accelerates heat loss through conduction.
- Heat loss due to temperature difference between the body and the environment.
- The rate of heat loss from the body is increased by being wet, by inactivity, dehydration, inadequate clothing, or shock.

Factors causing heat gain

- Gain of heat directly from the environment through radiation and conduction.
- Excessive fat deposits make it harder to lose the heat that is generated through activity.
- Heavy exercise, especially with excessive clothing.

**NOTE: The heat promoting centre is also called the "cold centre" and the heat losing centre is also called the "hot centre". We have used the terminology descriptive of the activities promoted by the centre in each case.*

Counteracting Heat Gain

Heat losing centre* in the hypothalamus monitors any rise in skin or core temperature above 37.5°C and coordinates responses that increase heat loss. These responses are mediated primarily through the **parasympathetic nerves** of the autonomic nervous system.

Sweating increases. Sweat cools by evaporation.

Muscle tone and **metabolic rate** are decreased. These mechanisms reduce the body's heat output.

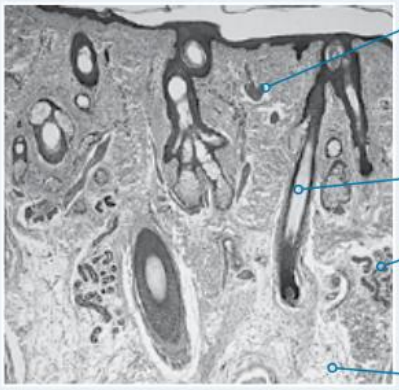
Blood flow to skin (**vasodilation**) increases. This increases heat loss.

Erector muscles of hairs relax to flatten hairs and decrease insulating air layer.

The skin and thermoregulation

Thermoreceptors in the dermis (probably free nerve endings) detect changes in skin temperature outside the normal range and send nerve impulses to the hypothalamus, which mediates a response. Thermoreceptors are of two types: **hot thermoreceptors** detect a rise in skin temperature above 37.5°C whereas the **cold thermoreceptors** detect a fall below 35.8°C. Temperature regulation by the skin involves **negative feedback** because the output is fed back to the skin receptors and becomes part of a new stimulus-response cycle.

Note that the thermoreceptors detect the temperature change, but the hair erector muscles and blood vessels are the effectors for mediating a response.



Cross section through the skin of the scalp.

Blood vessels in the dermis dilate (vasodilation) or constrict (vasoconstriction) to respectively promote or restrict heat loss.

Hairs raised or lowered to increase or decrease the thickness of the insulating air layer between the skin and the environment.

Sweat glands produce sweat in response to parasympathetic stimulation from the hypothalamus. Sweat cools through evaporation.

Fat in the subdermal layers insulates the organs against heat loss.

1. State two mechanisms by which body temperature could be reduced after intensive activity (e.g. hard exercise):

   (a) _____ (b) _____

2. Briefly state the role of the following in regulating internal body temperature:

   (a) The hypothalamus: _____

   (b) The skin: _____

   (c) Nervous input to effectors: _____

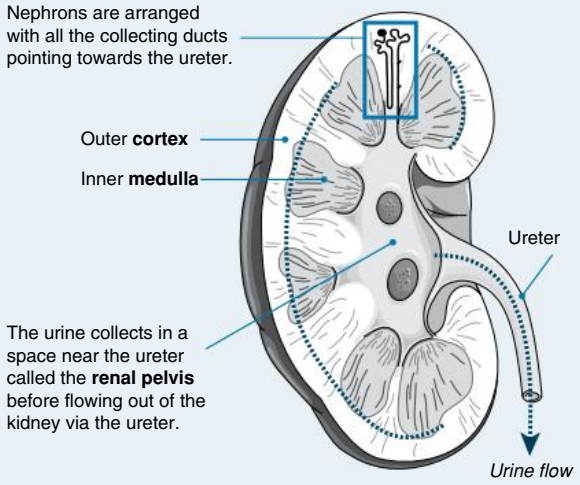
   (d) Hormones: _____

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68 The Physiology of the Kidney

Key Idea: The nephron is the functional unit of the kidney. It is a selective filter element, comprising a renal corpuscle and its associated tubules and ducts.

The functional unit of the kidney, the nephron, filters the blood and modifies that filtrate to produce an excretory fluid called urine. **Ultrafiltration**, i.e. forcing fluid and dissolved substances through a membrane by pressure, occurs in the first part of the nephron, across the membranes of the capillaries and the glomerular capsule. After formation of the initial filtrate, the **urine** is modified through **secretion** and tubular **reabsorption** according to body's need to retain or excrete water or ions.

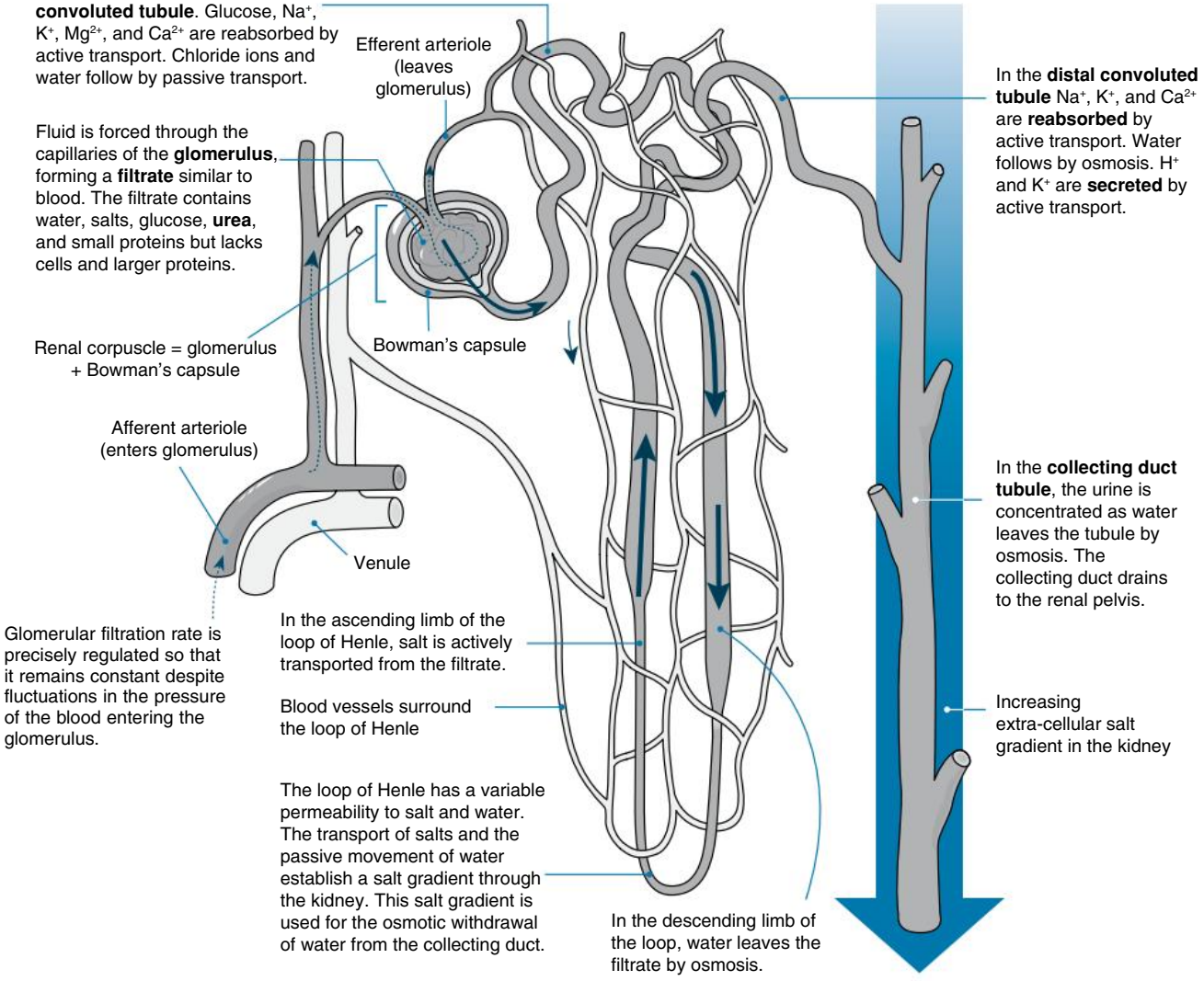


Internal Structure of the Human Kidney

Human kidneys are about 100-120 mm long and 25 mm thick. The functional unit of the kidney is the **nephron**. The other parts of the urinary system are primarily passageways and storage areas. The inner tissue of the kidney appears striated (striped), because of the orientation and alignment of the nephrons and blood vessels in the kidney tissue. It is this precise arrangement that makes it possible to fit in all the filtering units required.

Each kidney contains more than 1 million nephrons. By selectively filtering the blood plasma, the nephrons regulate blood composition and pH, and excrete wastes (e.g. urea) and toxins. The initial fluid is formed by **filtration** through the specialised epithelium of the glomerulus. This filtrate is then modified as it passes through the tubules of the nephron. The resulting **urine**, passes out the ureter.

Nephron Structure and Function



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Summary of Activities in the Kidney Nephron

Urine formation begins by **ultrafiltration** of the blood, as fluid is forced through the capillaries of the glomerulus, forming a filtrate similar to blood but lacking cells and proteins. The filtrate is then modified by secretion and **reabsorption** to add or remove substances (e.g. ions). The processes involved in urine formation are summarised below for each region of the nephron: glomerulus, proximal convoluted tubule, loop of Henle, distal convoluted tubule, and collecting duct.

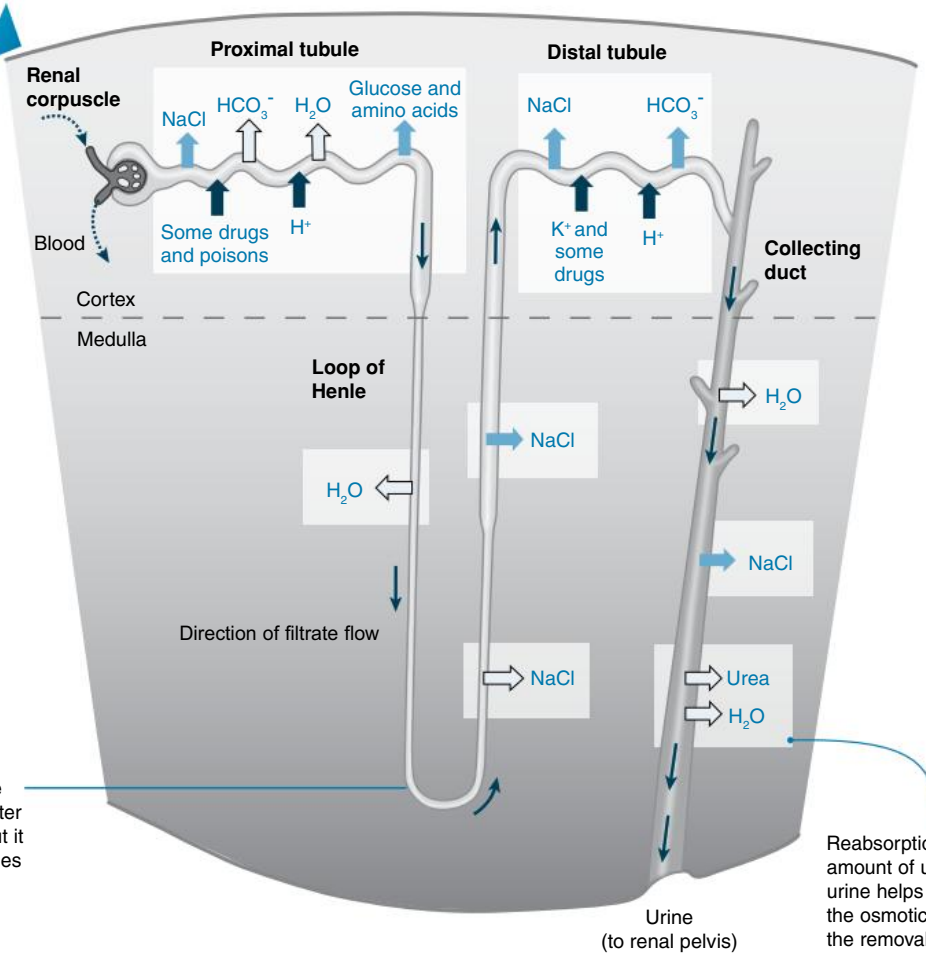


Filtrate

- H_2O
- Salts (e.g. NaCl)
- HCO_3^- (bicarbonate)
- H^+
- Urea
- Glucose, amino acids
- Some drugs

Reabsorption

- Active transport
- Passive transport
- Secretion (active transport)



The loop of Henle has varying permeability to salt and water. The transport of salts establishes and maintains the salt gradient across the medulla needed to concentrate the urine in the collecting duct. Water follows the salt out of the filtrate but it is transported away by the capillaries so that the high interstitial salt gradient is maintained.

Reabsorption of a small amount of urea from the urine helps to maintain the osmotic gradient for the removal of water.

1. (a) What is a **nephron**? _____

   (b) What is its role in excretion? _____

2. Explain the importance of the following in the production of urine in the kidney nephron:

   (a) Filtration of the blood at the glomerulus: _____

   (b) Active secretion: _____

   (c) Reabsorption: _____

   (d) Osmosis: _____

3. (a) What is the purpose of the salt gradient in the kidney? _____

   (b) How is this salt gradient produced? _____

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69 Control of Urine Output

Key Idea: Two hormones, antidiuretic hormone (ADH) and aldosterone, are involved in controlling urine output.

Variations in salt and water intake and environmental conditions contribute to fluctuations in blood volume and composition. The primary role of the kidneys is to regulate blood volume and composition (including the removal of nitrogenous wastes), so that homeostasis is maintained. This is achieved through varying the volume and composition of the urine. Two hormones, antidiuretic hormone (ADH) and aldosterone, are involved in the process.



Control of Urine Output

Osmoreceptors in the **hypothalamus** detect a fall in the concentration of water in the blood. They stimulate **neurosecretory cells** in the hypothalamus to synthesise and secrete the hormone ADH (antidiuretic hormone).

ADH passes from the hypothalamus to the posterior pituitary where it is released into the blood. ADH increases the permeability of the kidney collecting duct to water so that more water is reabsorbed and urine volume decreases.

Factors inhibiting ADH release

- Low solute concentration
  - High blood volume
  - Low blood sodium levels
- High fluid intake
- Alcohol consumption

ADH levels decrease → Water reabsorption decreases. Urine output increases.

Factors causing ADH release

- High solute concentration
  - Low blood volume
  - High blood sodium levels
- Low fluid intake
- Nicotine and morphine

ADH levels increase → Water reabsorption increases. Urine output decreases.

Factors causing release of aldosterone

Low blood volumes also stimulate secretion of aldosterone from the adrenal cortex. This is mediated through a complex pathway involving the hormone renin from the kidney.

Aldosterone → Sodium reabsorption increases, water follows, blood volume restored.

1. (a) **Diabetes insipidus** is a type of diabetes, caused by a lack of ADH. Based on what you know of the role of ADH in kidney function, describe the symptoms of this disease:

   _____

   _____

   (b) How might this disorder be treated? _____

   _____

2. Why does alcohol consumption (especially to excess) cause dehydration and thirst? _____

   _____

3. (a) What is the effect of aldosterone on the kidney nephron? _____

   (b) What is the net result of its action? _____

4. How do negative feedback mechanisms operate to regulate blood volume and urine output? _____

   _____

   _____

   _____

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LINK 68 LINK 51 WEB 69 KNOW

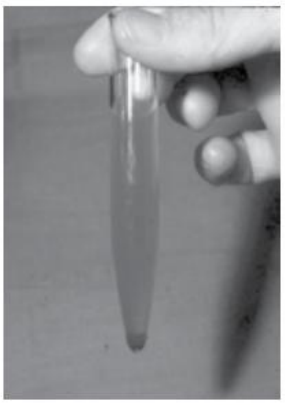
70 Urine Analysis

Key Idea: Urine analysis can be used to detect medical disorders, pregnancy, and the use of illegal drugs.

Urine analysis (**urinalysis**) is used as a medical diagnostic tool for a wide range of metabolic disorders. In addition, urine analysis can be used to detect the presence of illicit (non-prescription) drugs and for diagnosing pregnancy.

Diagnostic Urinalysis

A urinalysis (UA) is an array of tests performed on urine. It is one of the most common methods of medical diagnosis, as most tests are quick and easy to perform, they are non-invasive, and well understood diagnostically. A typical urinalysis usually includes a **macroscopic analysis**, a **dipstick chemical analysis**, in which the test results can be read as colour changes, and a **microscopic analysis**, which involves centrifugation of the sample and examination for crystals, blood cells, or microbial contamination.



MACROSCOPIC URINALYSIS

The first part of a urinalysis is direct visual observation. Normal, fresh urine is pale to dark yellow or amber in colour and clear.

Turbidity or cloudiness may be caused by excessive cellular material or protein in the urine. A red or red-brown colour could be from a food dye, eating fresh beets, a drug, or the presence of either haemoglobin or myoglobin. If the sample contained many red blood cells, it would be cloudy as well as red, as in this sample indicating blood in the urine.



DIPSTICK URINALYSIS

Commonly dipstick tests indicate:

Urine pH: normal range is 4.5-8.0.

Specific gravity: Normal is 1.002 - 1.035 Specific gravity measures the ability of the kidney to concentrate or dilute the urine over that of plasma.

Protein: Normal total protein excretion does not exceed 10 mg per 100 mL in any single specimen. More than 150 mg per day is defined as proteinuria.

Glucose: Less than 0.1% of glucose filtered by the glomerulus normally appears in urine. Excess sugar in urine generally indicates diabetes mellitus.

Ketones: Ketones in the urine result from diabetic ketosis or some other form of calorie deprivation (starvation).

Nitrite: Nitrites indicate that bacteria may be present in significant numbers.

Leucocyte esterase: A positive leucocyte esterase test results from the presence of whole or lysed white blood cells (indicating infection).

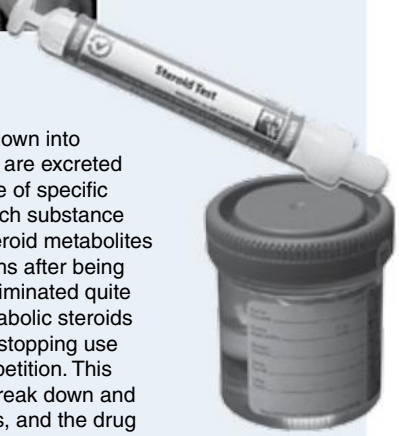
Testing For Anabolic Steroids

Anabolic steroids are synthetic steroids related to the male sex hormone **testosterone** (right). They work by increasing protein synthesis within cells, causing tissue, especially skeletal muscle, to build mass. They are used legitimately in medicine to stimulate bone growth and appetite, induce male puberty, and treat chronic wasting conditions. Misuse of anabolic steroids can result in elevated blood pressure, cardiovascular disease, and alteration of cholesterol ratios.





Steroids increase muscle mass and physical strength, and are used illegally by some athletes to gain an unfair advantage over competitors. Anabolic steroid use is banned by most major sporting bodies, but many athletes continue to use them. Athletes are routinely tested for the presence of performance enhancing drugs, including anabolic steroids.



Anabolic steroids break down into known metabolites which are excreted in the urine. The presence of specific metabolites indicates which substance has been used. Some steroid metabolites stay in the urine for months after being taken, while others are eliminated quite rapidly. Athletes using anabolic steroids can escape detection by stopping use of the drugs prior to competition. This allows the body time to break down and eliminate the components, and the drug use goes undetected.

1. Why is **urinalysis** a frequently used diagnostic technique for many common disorders? _____

2. Explain why the pH of normal urine (4.5-8.0) is much more variable than the pH of the blood (pH 7.35-7.45):

3. What might each of the following abnormalities in a urine sample indicate to a doctor?

   (a) Cloudy, red colour: _____ (b) Positive leucocyte esterase test: _____

4. Why might an athlete, who is exploiting illegal drugs, withhold them for a period of time before competition?

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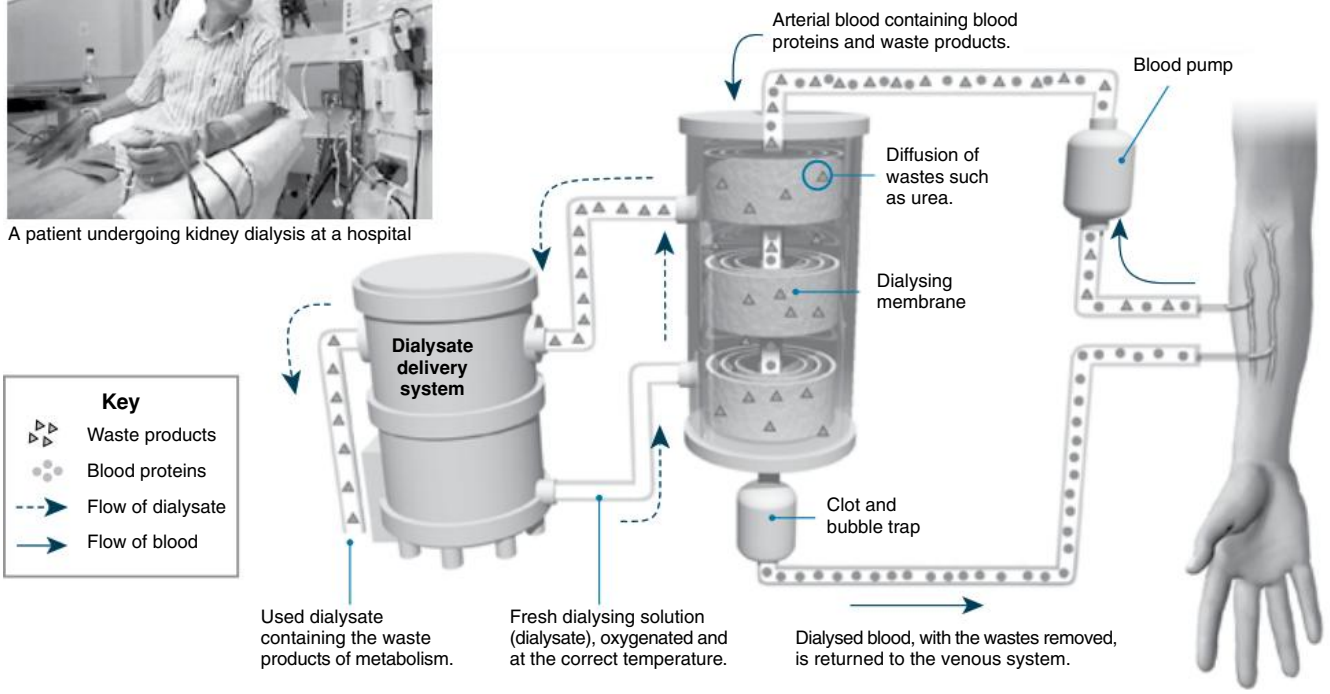
71 Renal Dialysis

Key Idea: A kidney dialysis machine acts as an artificial kidney, removing waste from the blood when the kidneys fail. When the kidneys do not function properly, waste products build up in the body, and medical intervention is required to correct the problem. A dialysis machine removes wastes from the blood. It is used when the kidneys fail, or when blood acidity, urea, or potassium levels increase above normal. Blood flows through a system of tubes composed of partially permeable membranes. Dialysis fluid (dialysate) has a composition similar to blood except that the concentration of wastes is low. It flows in the opposite direction to the blood on the outside of the dialysis tubes. Consequently, waste products like urea diffuse from the blood into the dialysis fluid, which is constantly replaced. The dialysis fluid flows at a rate of several 100 cm^3 per minute over a large surface area Dialysis can be ongoing, or can be used to allow the kidneys to rest and recover from injury, the effects of drugs or other metabolic disturbance.



A patient undergoing kidney dialysis at a hospital

Principles of Kidney Dialysis



1. In kidney dialysis, explain why the dialysing solution is constantly replaced rather than being recirculated:

2. Explain why ions such as potassium and sodium, and small molecules like glucose do not diffuse rapidly from the blood into the dialysing solution along with the urea:

3. Why does the urea pass from the blood into the dialysing solution?

4. Describe the general transport process involved in dialysis:

5. Why does the dialysing solution flow in the **opposite** direction to the blood?:

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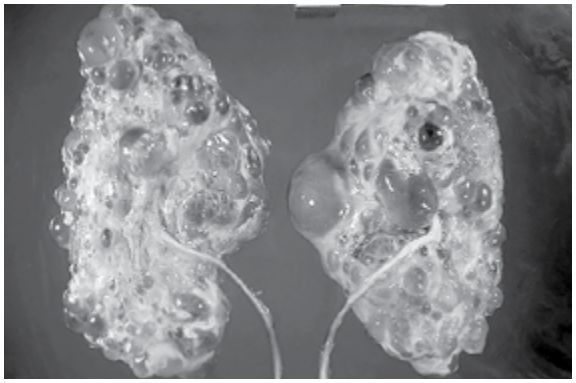
72 Kidney Transplants

Key Idea: A kidney transplant, where a healthy kidney from one person is transplanted into another, is a procedure for people with complete kidney failure.

Kidney failure (renal failure) arises when the kidneys fail to function adequately and filtrate formation decreases or stops. In cases of renal failure, normal blood volume levels and electrolyte balances are not maintained, and waste products build up in the body. Kidney failure (renal failure) may come on suddenly (acute) or develop over a long period of time (chronic). Recovery from acute kidney failure is possible, but chronic kidney damage cannot be reversed. When the damage is extensive, a kidney transplant is required.

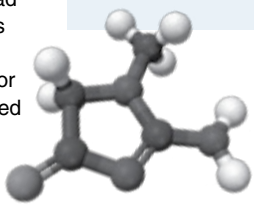
Renal Failure

Kidney (renal) failure is indicated by levels of **serum creatinine**, as well as by kidney size on ultrasound and the presence of anaemia (chronic kidney disease generally leads to anaemia and small kidneys). Creatinine is a break-down product of creatine phosphate in muscle, and is usually produced at a fairly constant rate by the body (depending on muscle mass). It is chiefly filtered from the blood by the kidneys, although a small amount is actively secreted by the kidneys into the urine. Blood creatinine levels only rise when there is marked damage to functioning nephrons.



CDC

Acute renal failure (ARF) is characterised by a drop in urine production (<400 mL per day), and commonly arises because of low blood volume (blood loss), dehydration, or widespread infection. In contrast, **chronic renal failure**, which develops over months or years, is commonly the result of poorly controlled diabetes, poorly controlled high blood pressure, or **polycystic kidney disease**, a genetic disorder characterised by the growth of numerous cysts in the kidneys (above).



Creatinine

Creatinine levels in both blood and urine is used to calculate the creatinine clearance (CrCl), which reflects the glomerular filtration rate (GFR). The GFR is a clinically important measurement of renal function and more accurate than serum creatinine alone, since serum creatinine only rises when nephron function is very impaired.

Kidney Transplants



Transplantation of a healthy kidney from an organ donor is the preferred treatment for end-stage kidney failure. The organ is usually taken from a person who has just died, although kidneys can also be taken from living donors. The failed organs are left in place and the new kidney transplanted into the lower abdomen. If recipients comply with medical requirements (e.g. correct diet and medication) more than 85% of kidney transplants are successful.

There are two major problems associated with kidney transplant technology: lack of donors and tissue rejection. Cells from donor tissue have different antigens to those of the recipient, and are not immuno-compatible. Tissue-typing and the use of immunosuppressant drugs helps to decrease rejection rates. In the future, **xenotransplants** of genetically modified organs from other species may help to solve both the problems of supply and immune rejection.

1. Distinguish between acute and chronic renal failure and contrast their causes: _____

2. (a) Why would a rise in blood (serum) levels of creatinine indicate a failure of nephron function? _____

   (b) Why is creatinine clearance a more accurate indicator of renal function than a serum creatinine test alone?

3. Describe some of the advantages and disadvantages of kidney transplantation over a life-time of kidney dialysis:

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73 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its definition, as identified by its preceding letter code.

apoptosis

autocrine signalling

biological transducer

cyclic AMP

diabetes mellitus

effector

endocrine signalling

glucagon

homeostasis

hormone

insulin

mechanoreceptor

negative feedback

nervous system

neurohormone

neurotransmitter

paracrine signalling

pheromone

photoperiodism

plant growth regulators

positive feedback

receptor

response

second messenger

signal transduction

stimulus

synapse

A A chemical messenger that relays signals between a neurone and another cell in response to a nerve impulse.

B A hormone produced and released by modified neurones.

C A sensory receptor that responds to the stimulus modality of mechanical pressure or distortion.

D Cell signalling in which a cell secretes and reacts to its signal molecules.

E The gap between neighbouring neurones or between a neurone and an effector.

F The process of programmed cell death in multicellular organisms, in which the cell goes through a series of orderly chemical events leading to death

G A detectable change in the internal or external environment that is capable of eliciting a response in an organism.

H A molecule that relays signals from receptors on the cell surface to target molecules inside the cell.

I A hormone, secreted by the pancreas, that raises blood glucose levels.

J In cell signalling, a molecule that binds to a protein and alters its activity. In a neural pathway, a muscle or gland capable of responding to a nerve impulse.

K A hormone, secreted by the pancreas, that lowers blood glucose levels.

L An organ system comprising a network of specialised cells or neurones, which coordinates responses and transmits signals between different parts of the body.

M A chemical messenger released into the external environment which acts as a signal to another animal.

N Signalling molecules, produced within plants, that regulate cellular processes in target cells. Also known as plant hormones.

O Signalling that involves hormones released by ductless endocrine glands and carried through the body by the circulatory system to target cells.

P A destabilising mechanism in which the output of the system causes an escalation in the initial response.

Q Signalling involving a chemical messenger whose effect is only exerted at short range on neighbouring cells.

R In cell signalling, a molecule (usually) on the cell surface, which receives chemical signals originating from outside the cell. In a neural pathway, a sensory structure that responds to a stimulus in the internal or external environment of an organism.

S A mechanism by which a mechanical or chemical stimulus to a cell is converted into a specific cellular response.

T A mechanism in which the output of a system acts to oppose changes to the input of the system. The net effect is to stabilise the system and dampen fluctuations.

U Regulation of the internal environment to maintain a stable, constant condition.

V An important intracellular signal transduction molecule, derived from ATP. The second messenger in many signal transduction pathways.

W Biological structures, such as sensory receptors, that convert one type of energy (e.g. light) into another (e.g. electrical impulse).

X A condition in which the blood glucose level is elevated above normal levels, either because the body doesn't produce enough insulin, or because the cells do not respond to the insulin that is produced.

Y A signalling molecule, produced by an endocrine gland, that is secreted into the blood and affects the metabolism of specific target cells.

Z The behaviour occurring as a result of an external or internal stimulus.

AA The physiological response to light in plants mediated by phytochrome.

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QLD As required
VCE Unit 3 (2)
WA ATAR Unit 4
SA N/A

The Nature of Pathogens

Key terms

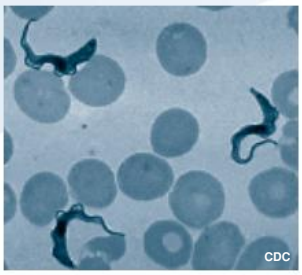
antibiotic
antigen
antiviral
bacteria
disease
fungi
immunisation
infectious disease
isolation
Oomycetes
parasite
pathogen
prion
protist
protozoan
quarantine
vector (for disease)
virulence
virus
water mould
zoonoses

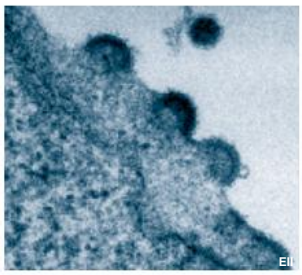
The nature of pathogens

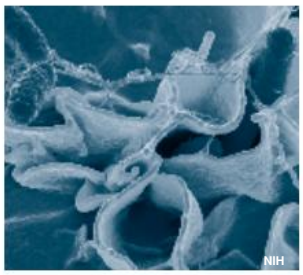
Learning aims and skills

Activity number

☐	1	Describe the nature of pathogens and explain their role in infectious disease. Explain how pathogens act as antigens. Identify types of pathogens, including bacteria, fungi, viruses, protists, and prions by their structural characteristics.	74 77 80 81 83 87
☐	2	Describe modes of transmission of disease, including the role of insect vectors. Use an example to describe the characteristics of zoonoses.	75
☐	3	Describe and explain the factors involved in the spread of a disease in a population, including the growth of the pathogen population, the density of the host population, mode of transmission, and reservoirs of infection.	74 76







Pathogens and disease

Learning aims and skills

Activity number

☐	4	For chosen examples as appropriate, describe the life cycle of a pathogen, the disease it causes, method of invading the host, and mode of transmission. Comment on how these features of the pathogen determine its success. Choose from the examples below for each of the pathogen groups.	78-88
☐	5	**Bacterial pathogens**: Diseases include tuberculosis, cholera, tetanus, and crown gall in plants.	77-79
☐	6	**Fungal pathogens**: Diseases include amphibian chytrid fungus disease.	80
☐	7	**Protistan pathogens**: Diseases include malaria (protozoan) and *Phytophthora* dieback (Oomycetes or water moulds, not to be confused with fungal moulds).	81 82
☐	8	**Viral pathogens**: Diseases include HIV/AIDS, influenza, Ross River virus, sac brood in honeybees, and Australian bat Lyssavirus.	83-87
☐	9	**Prion pathogens**: Diseases include variant Creutzfeldt–Jakob disease (vCJD, kuru, BSE, and scrapie.	88

Responding to pathogens

Learning aims and skills

Activity number

☐	10	Explain how the transmission and spread of diseases is assisted by the ease with which organisms can move between different regions and parts of the globe.	76
☐	11	Relate the nature of the pathogen to the strategies used to control and treat the disease it causes. Include reference to the role of isolation, quarantine, immunisaton, and medications (antibiotics and antivirals). Evaluate the evidence for the role of immunisation programmes and the role of herd immunity in managing disease outbreaks.	89 90



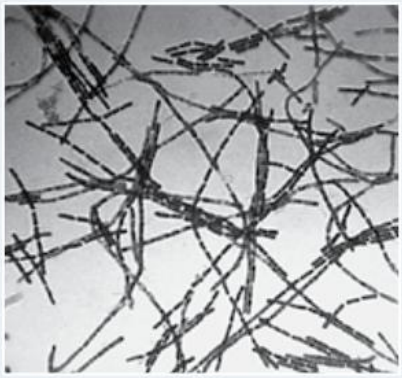
74 Infection and Disease

Key Idea: Pathogens are infectious agents that spread between hosts and cause disease. They carry or produce antigens that can be identified by the host as foreign.

Pathogens are agents of infectious disease. They, or the toxins they produce, act as antigens (substances that are recognised as foreign by the host's immune system) and promote an immune response. The goal of a pathogen is to multiply and spread between hosts before being detected by the host's immune system. Many pathogens have highly effective modes of transmission and so are highly infectious.

Types of Pathogen

CDC



Bacillus anthracis bacterium causes anthrax. The anthrax bacillum can form long-lived spores.

Bacteria: All bacteria are prokaryotes. Many bacteria are useful, but the relatively few pathogenic species are responsible for enormous social and economic cost. This is especially so since the rise in incidence of antibiotic resistance. Bacterial diseases can be treated with antibiotics or prevented through vaccination.

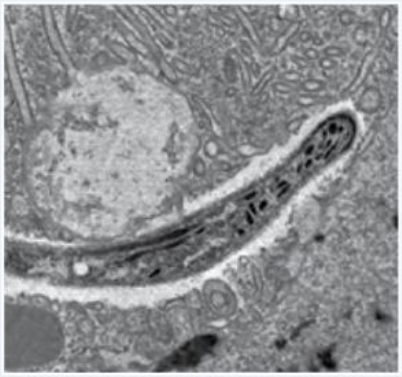
CDC



Photo: Bangladeshi girl with smallpox (1973). Smallpox was eradicated from the country in 1977.

Viruses are obligate intracellular parasites and need living host cells in order to multiply. Antibiotics, which target bacterial cells only, are ineffective against viruses, but viral diseases can be effectively controlled, or even eradicated, through vaccination programmes.

Photo: Ute Frevert



Malaria sporozoite moving through gut epithelia. The parasite is carried by a mosquito vector.

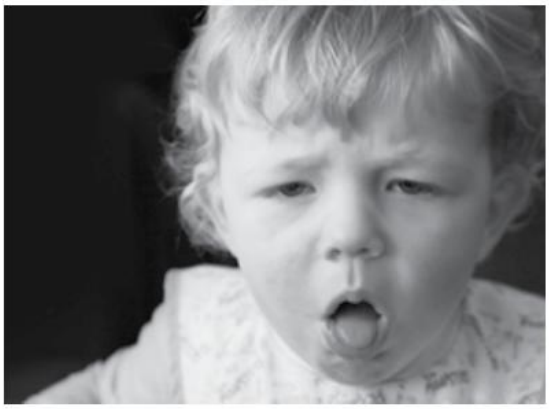
Eukaryotic pathogens include fungi, algae, protozoa, and parasitic worms. Many are highly specialised parasites with a number of hosts, e.g. the malaria parasite has a mosquito and a human host. Certain stages in its life cycle reside within the host's cells, making them (like viruses) hard to treat.

Pathogen Success and Failure

In general, a host's immune system quickly recognises an invading pathogen as foreign, usually on the basis of its surface proteins or the toxins it produces. This recognition triggers an immune (defensive) response in the host that destroys the pathogen. To be successful, a pathogen must avoid the host's immune system for long enough to infect a new host.

Different pathogens use different strategies to avoid detection and infect other hosts. They may multiply rapidly and spread before the immune system can react (e.g. common cold, right) or they may produce a slower chronic infection that is more difficult for the immune system to overcome (e.g. HIV/AIDS).

Pathogens that kill the host before being able to spread are not successful. In high density populations, diseases with a high mortality (death rate) such as Bubonic plague, Ebola, and smallpox, can spread successfully (witness the 2014 Ebola outbreaks in western African countries). However, in low density areas, pathogens that kill their host quickly have poor transmission, as too few hosts can be infected during the infectious period.



One method a pathogen can use to spread between hosts is to cause an irritation in the throat that produces a cough. The cough spreads infected droplets to other hosts.

1. (a) What is a pathogen? _____

   _____

   (b) Identify at least four different types of pathogens: _____

   _____

2. What must a pathogen do to be successful? _____

   _____

   _____

3. Explain why some outbreaks of infectious disease "burn out" quickly: _____

   _____

   _____

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75 Transmission of Disease

Key Idea: Infectious disease can spread rapidly within and between regions given the right conditions.

The human body, like that of other large animals, is constantly exposed to a wide range of potential parasites and pathogens. Transmission and spread of a pathogen depends on its rate of growth, the density of the host population, the mobility of the host population, and the mode of transmission. The transmission of infectious diseases can be virtually eliminated by observing hygienic practices, e.g. chlorinating drinking water and by providing adequate sanitation.

Transmission and Spread



Most pathogens, once inside the body, multiply rapidly, producing symptoms and making the host infectious within a few days. Others take longer to present symptoms. The infectious period can last from a few days to weeks, but in some cases the host may be infectious for long periods of time.

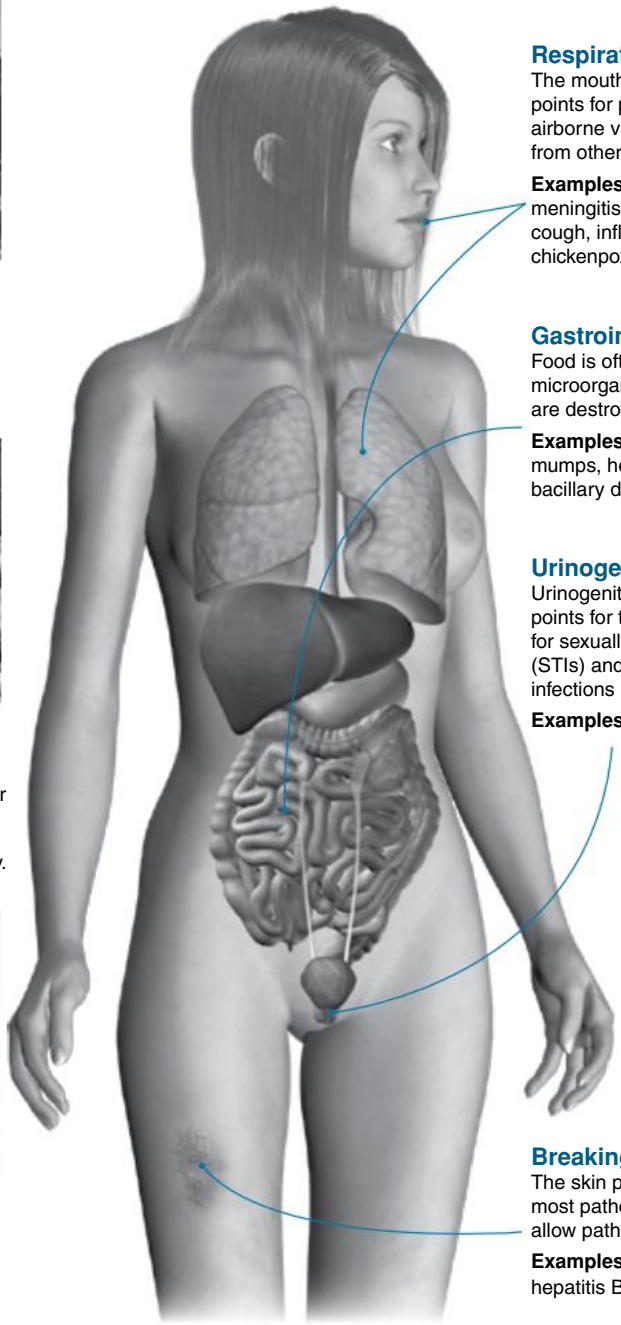


Human cities can contain millions of people, often living very closely together. In these congested conditions, infectious diseases can spread rapidly, especially if sanitation or personal hygiene is poor, or if seasonal weather produces conditions favourable to spread of the pathogen. High speed transport can help spread a pathogen around a region very quickly.



The mode of transmission affects how quickly a pathogen spreads. Direct person to person contact (i.e. touching) is a slower method of spreading, while spreading via mucus droplets coughed into the air or by animal vectors can help a pathogen spread quickly and widely.

Portals of Entry



Respiratory tract

The mouth and nose are major entry points for pathogens, particularly airborne viruses, which are inhaled from other people's expelled mucus.

Examples: diphtheria, meningococcal meningitis, tuberculosis, whooping cough, influenza, measles, rubella, chickenpox.

Gastrointestinal tract

Food is often contaminated with microorganisms, but most of these are destroyed in the stomach.

Examples: cholera, typhoid fever, mumps, hepatitis A, poliomyelitis, bacillary dysentery, salmonellosis.

Urinogenital openings

Urinogenital openings provide entry points for the pathogens responsible for sexually transmitted infections (STIs) and other opportunistic infections (i.e. thrush).

Examples: gonorrhoea, HIV.

Breaking the skin surface

The skin provides an effective barrier to most pathogens, but cuts and abrasions allow pathogens to penetrate.

Examples: tetanus, gas gangrene, hepatitis B, rabies, malaria, and HIV.

1. Why can disease spread quickly in congested human cities? _____

2. Why would transmission by direct touch be slower than transmission by coughing or sneezing? _____

KNOW LINK 76 LINK 90

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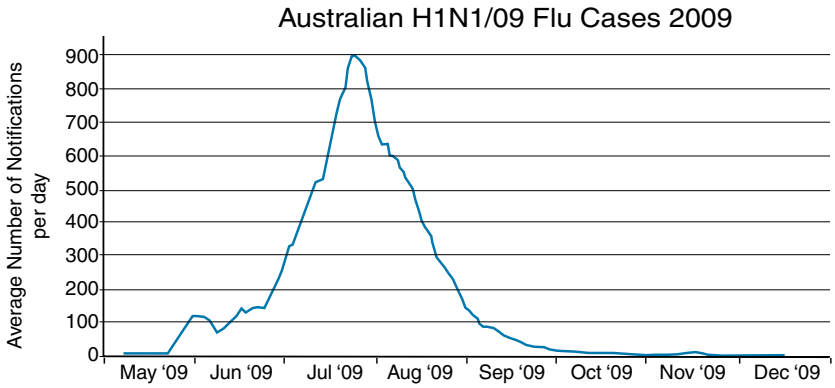
76 Patterns of Disease

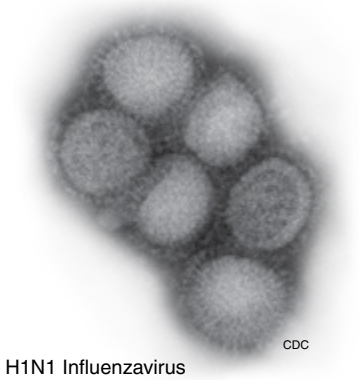
Key Idea: Studying the prevalence and spread of a disease gives insights into its origins and how to combat it.

Diseases present at constant low levels in a population or region are known as **endemic** diseases. Occasionally there may be a sudden increase in the **prevalence** of a particular disease. On a local level this is known as an **outbreak**. When an infectious disease spreads rapidly through a nation and affects large numbers of people it is called an **epidemic**. On rare occasions a new kind of disease will appear and spread to other countries. The rapid spread of a disease throughout the world is a **pandemic**. Examples of pandemic diseases include tuberculosis, HIV/AIDS, and influenza.

Epidemiologists gather data on the number of infected people (**morbidity**) and the number of people that have died (**mortality**) within a population. These data help to establish the **incidence** (number of new cases per unit time) and **prevalence** (number of infected people expressed as a proportion of the population) of the disease in the population at any given time. **Aetiology** is the study of the cause of a disease. It can assist in pinpointing the origin of new diseases, such as the H1N1/09 influenza virus (swine flu).

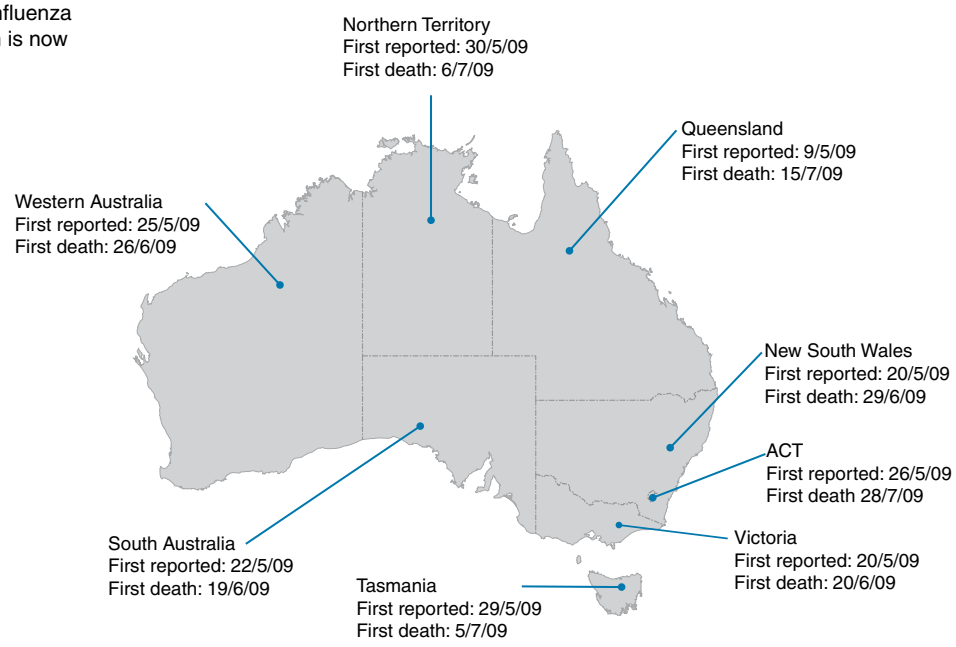
The H1N1/09 subtype of the influenza virus (formerly known as swine flu), crossed the species barrier to humans in Mexico and from there spread to the rest of the world. Cases in Australia were first reported in May and quickly rose to a peak in late July. Total cases for 2009 were over 37,000. The 2009 H1N1/09 pandemic was the first influenza pandemic of the 21st century. The strain is now included in the annual flu vaccination.





H1N1 Influenzavirus

Spread of H1N1 Flu Across Australia



1. (a) Why is H1N1/09 classed as a pandemic?_____

(b) What is the most likely origin of H1N1/09 spread in Australia? Explain your answer:_____

(c) Suggest likely reasons for the rapid fall-off in the incidence of H1N1/09 in Australia after August 2009:

2. Why is it important to establish the **incidence** of a disease when it begins to spread through a community?

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LINK 103 LINK 90 KNOW

77 Bacterial Diseases

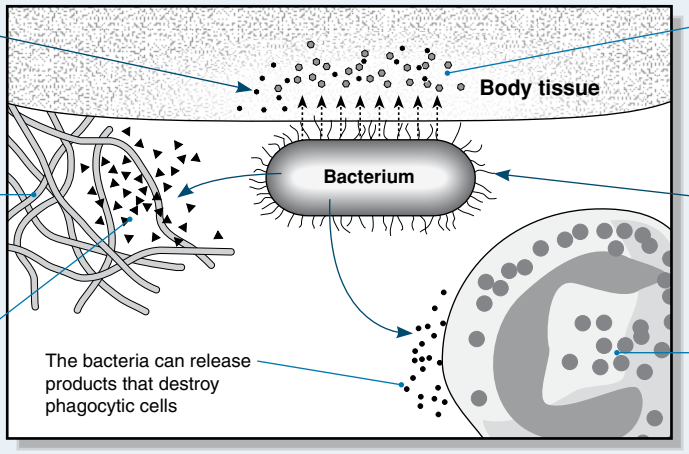
Key Idea: Pathogenic bacteria are responsible for some of the world's most devastating diseases of plants and animals. Relatively few of the world's bacterial species cause disease. Those that do (the pathogenic bacteria), have a range of adaptations that enable them to penetrate the defences of a host and cause infection (below). Bacterial diseases are commonly transmitted through food, water, air, or by direct contact. The natural source of infection of a disease varies from species to species, ranging from humans and other organisms, to sewage or contaminated water. Much of our control of bacterial disease is achieved through identifying reservoirs of infection and limiting the routes of transmission.

How Bacteria Invade a Host's Tissues

Toxins: Bacterial toxins can act locally to promote bacterial invasion (e.g. the enzymes that degrade collagen), or they may have cytotoxic activity and destroy cells directly.

Fibrin: Fibrous threads of protein are deposited when blood clots. This action by the host effectively limits the movement of pathogens in infected areas.

Enzymes are released that break down fibrin, allowing the bacteria greater freedom of movement.

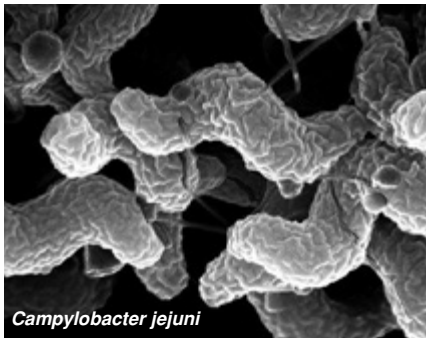


The bacterium releases enzymes that degrade the connective tissue of the host, allowing the spread of infection.

Fimbriae are fine, threadlike extensions from the bacterial cell. The bacteria use them to attach to the mucous membranes and directly attack the host tissues.

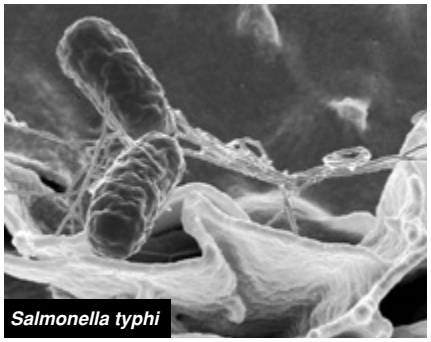
Phagocyte: These white blood cells are very effective in identifying and destroying foreign cells such as pathogens.

Methods of Bacterial Transmission


USDA
Campylobacter jejuni

Foodborne bacterial diseases

Bacterial foodborne illnesses are caused by consuming food or beverages contaminated with bacteria or their toxins. Examples include *Salmonella* food poisoning and *Campylobacter* infection. Symptoms of bacterial food poisoning include fever, abdominal cramps, and diarrhoea. Some are associated with consuming raw or undercooked poultry.


NIH
Salmonella typhi

Waterborne bacterial diseases

Waterborne bacterial pathogens are responsible for a number of serious diarrhoeal illnesses, including typhoid and cholera. Transmission of these diseases is usually through faecal contamination of drinking water. The fever and diarrhoea associated with such diseases is responsible for hundreds of thousands of deaths annually in countries with poor sanitation.


CDC

Airborne bacterial diseases

Airborne pathogens are transmitted on dust particles or droplets when people cough, sneeze, or exhale. Vaccination against certain airborne bacteria has been highly successful. Whooping cough (above) is a potentially fatal respiratory disease caused by the bacterial pertussis toxin. The prevalence of this disease has declined dramatically following the introduction of immunisation programmes.

1. Describe the specific adaptations of bacteria that contribute to their ability to cause disease:

2. Why is immunisation often the best option for controlling airborne bacterial diseases?

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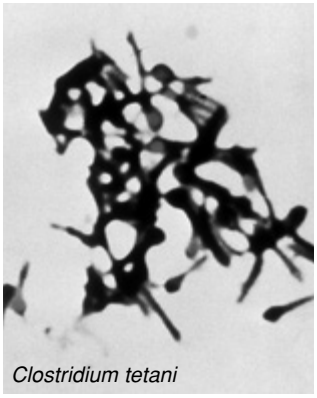
Tetanus

Tetanus is a condition characterised by prolonged, strong contractions of the skeletal muscles. It is caused by the toxin tetanospasmin, produced by the bacteria *Clostridium tetani*. Symptoms of tetanus begin with muscle spasms in the jaw (hence the common name "lockjaw") and spread through the chest, back and rest of the body. The spasms can affect breathing and swallowing and can be fatal in unvaccinated people. *Clostridium tetani* is an anaerobic, gram positive bacterium commonly found in the soil. Infection is usually through a puncture wound that becomes contaminated with soil or dirt. Tetanus can be prevented by vaccination. Booster shots need to be taken around every 10 years as the vaccine's effectiveness declines over time.

Plant Crown Gall

Crown gall is a tumour-like growth in plants. It is caused by the soil bacterium *Agrobacterium tumefaciens* (also known as *Rhizobium radiobacter*). The gall is produced when the bacterium transfers a circular piece of DNA (the tumour inducing or *Ti* plasmid) to the plant cell. *Agrobacterium tumefaciens* is of particular use in biotechnology as it can be used to insert new genes into plants for genetic modification. The cells making up the gall contain genes carried on the *Ti* plasmid. The gall itself can grow quite large, depending on the plant infected. As the disease progresses, the plant may grow poorly and eventually die as its water transport systems are cut off by the growth of the gall.


CDC


CDC *Clostridium tetani*


MSU *Agrobacterium*


USDA

3. Why is it not good practice to chop vegetables on the same chopping board as is used to prepare raw chicken?

4. Why are there often outbreaks (or risks of outbreak) of bacterial diseases such as typhoid and cholera after large scale natural disasters, such as large scale earthquakes or tsunamis?

5. How can simple measures, such as washing hands before eating, reduce the incidence of bacterial diseases?

6. Explain why *Agrobacterium tumefaciens* is of particular interest to scientists:

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78 Cholera

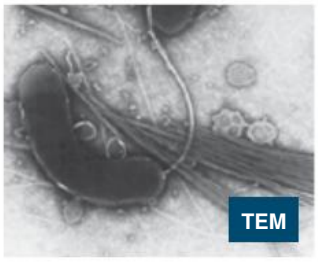
Key Idea: Cholera is a bacterial disease that causes severe diarrhoea and is spread by drinking contaminated water.

Cholera is an acute intestinal infection caused by the bacterium *Vibrio cholerae*. Infection results in severe diarrhoea, which may be fatal. Cholera is spread mainly by consuming contaminated drinking water or food. The disease can be prevented by hygienic disposal of human faeces, providing an adequate supply of safe drinking water, safe food handling and preparation, and effective general hygiene (e.g. hand washing with soap).

Cholera and Diarrhoea

The cholera bacterium (below) produces an **enterotoxin**, which increases the permeability of the intestinal epithelium to chloride ions and results in copious, painless, watery diarrhoea that can lead to severe dehydration, electrolyte imbalance, kidney failure, and death within hours if untreated.

Cholera is treated with **oral rehydration salts** (ORS) containing glucose and electrolytes, which are taken up by active transport by the intestinal cells. This reverses the osmotic gradient and allows blood volume to be restored.



Dartmouth Electron Microscope Facility

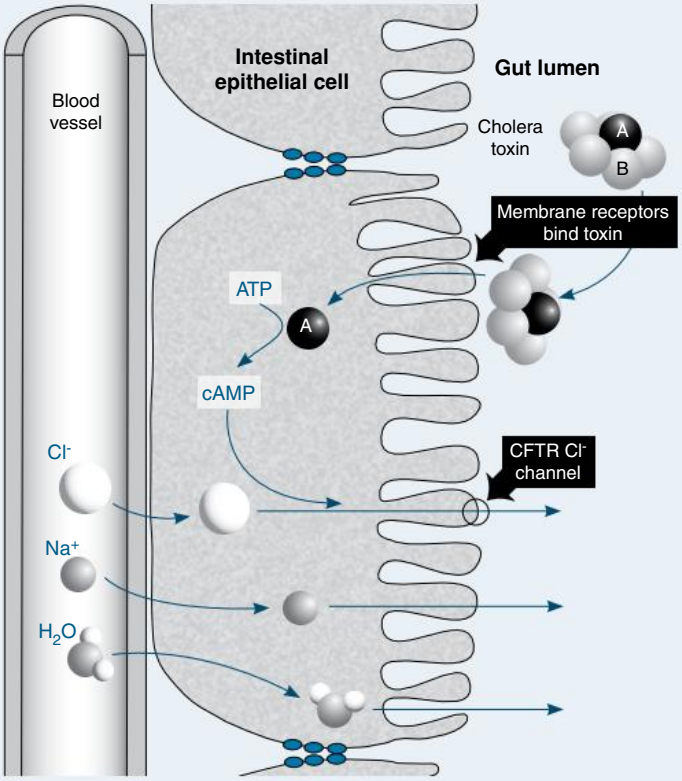
CDC



Roger LeMoyne UNICEF (www.unicef.org.nz)

A woman in Sudan learns to use ORS (Unicef)

Mechanism of Cholera-Induced Dehydration



Cholera toxin contains two subunits (A and B), which bind to receptors on the intestinal cell membrane. Subunit A enters the cell and activates pathways that cause the production of cAMP. cAMP opens transport (CFTR) channels, causing chloride ions (Cl^-) to leave the cell and enter the gut lumen (cavity). Sodium follows chloride down its electrochemical gradient, and water follows the salt down its osmotic gradient. These are replaced from the blood, causing a drop in blood volume (dehydration).

1. How do the symptoms of cholera relate to the transmission and success of the pathogen? _____

2. How does the cholera toxin cause a loss of water into the gut lumen? _____

3. (a) How are the symptoms of cholera treated? _____

   (b) How does this treatment work? _____

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79 Tuberculosis

Key Idea: Tuberculosis is a contagious disease affecting the lungs, causing scarring of the lung tissue.

Tuberculosis (TB) is a contagious disease caused by the bacterium *Mycobacterium tuberculosis* (MTB). TB most commonly affects the lungs as **pulmonary TB**. Presence of the disease is indicated on a chest X-ray by opaque areas and large thick walled cavities in the lungs resulting from bacterial damage. TB affects millions globally and increasing drug resistance is contributing to its impact. Effective treatment requires a prolonged regime of antibiotics, and poor patient compliance with treatment contributes to its persistence. At the current rate of TB spread, the WHO anticipate that approximately 1 billion people will be newly infected and 36 million will die from the disease between 2002 and 2020.

Pulmonary Tuberculosis

Infection and transmission

TB is spread through the air when infectious people cough, sneeze, talk, or spit. Transmission does not require a large inoculum; a person needs to inhale only a small number of *Mycobacterium tuberculosis* (MTB) to be infected. Left untreated, each person with active TB will infect 10-15 people every year. Those infected do not necessarily become ill because the immune system can 'wall off' the MTB which then lie dormant, protected by a thick waxy coat. When the immune system is weakened, the chance of becoming ill is much greater.

Symptoms

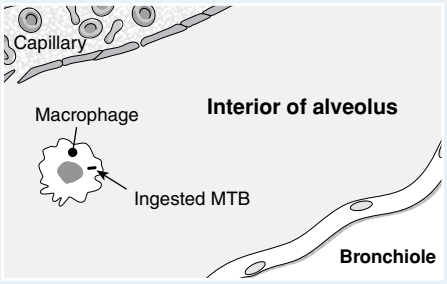
TB usually affects the lungs, but it can also affect other parts of the body, such as the brain, the kidneys, and the spine. The specific symptoms of pulmonary TB of include coughing, chest pain, and coughing up blood. The bacteria can spread from the bronchioles to other body systems, where the symptoms depend on the part of the body that is affected.

Effect on lung function

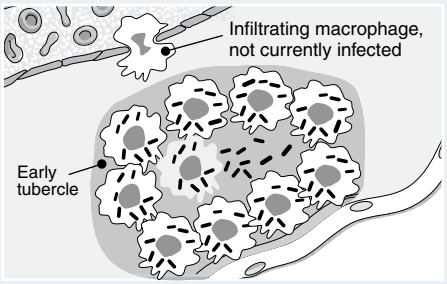
When MTB is inhaled, bacilli reach the lungs, where they are ingested by an alveolar macrophages. Usually the macrophages destroy the bacteria, but if they do not, the bacilli are hidden from the immune system and survive and multiply within the macrophages. More macrophages are attracted to the area and a tubercle forms. The disease may become dormant or the tubercle may rupture, releasing bacilli into the bronchioles (diagram panel, right). Affected tissue is replaced by scarring (fibrosis) and cavities filled with cheese-like white necrotic material. During active disease, some of these cavities are joined to the air passages. This material can be coughed up. It contains living bacteria which can pass on infection.

Stages in TB Infection

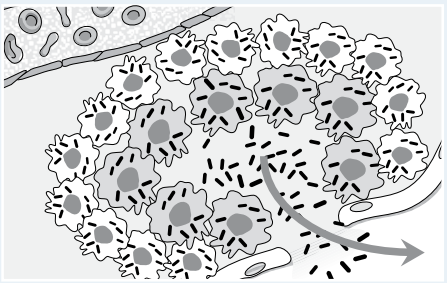
The series below illustrates stages in MTB infection.



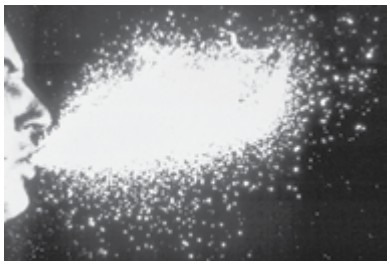
MTB enter the lung and are ingested by macrophages (phagocytic white blood cells).



The multiplying bacteria cause the macrophages to swell and rupture. The newly released bacilli infect other macrophages. At this stage a tubercle may form and the disease may lie dormant.



Eventually the tubercle ruptures, allowing bacilli to spill into the bronchiole. The bacilli can now be transmitted when the infected person coughs.



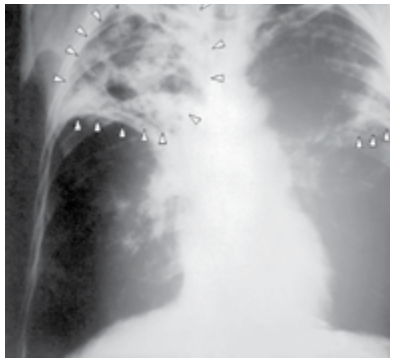
CDC

TB is spread by droplet transmission.



CDC: Janice Haney Carr

Mycobacterium tuberculosis (MTB)



CDC

Above: X-ray of lungs affected by pulmonary TB. The white triangles indicate areas where the airspaces of the lung are congested with fluid, and the dark arrows indicate a cavity, from which infective material is coughed up. Surface area for gas exchange is reduced and lung function is adversely affected.

1. Describe how the TB pathogen causes infection and spreads between hosts: _____

2. How do features of the pathogen's biology contribute to its success? _____

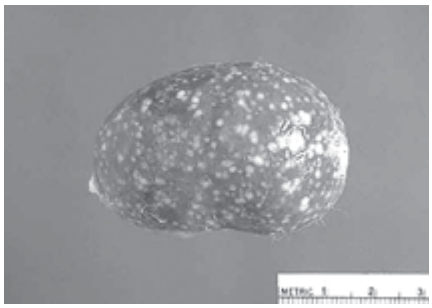
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80 Fungal Diseases

Key Idea: Pathogenic fungi are rare in animals, but they can cause infections that are long lasting and difficult to treat.

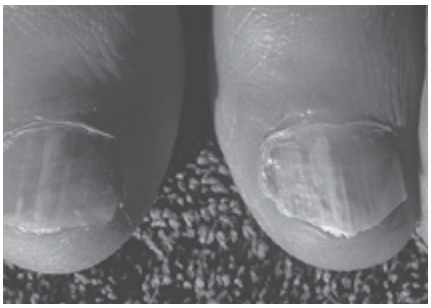
All fungi are heterotrophic, requiring organic compounds for energy and carbon. They may be parasitic or saprotrophic, obtaining nutrition by the extracellular digestion of living or dead organic matter. Very few fungi are pathogenic to animals, although thousands of fungal species are plant pathogens. They spread by spores and the infections they cause are generally **chronic** (long-lasting) infections because fungi grow relatively slowly. Fungal diseases are categorised into three broad groups (below), the most common being superficial infections of the skin. Of great concern recently is the spread of a fungal pathogen in amphibian populations. Amphibian chytrid fungus disease has been linked to the dramatic decline in amphibian populations globally. Amphibians rely on their skin for osmoregulation and oxygen uptake, so they are particularly vulnerable to pathogens that compromise its integrity.

Types of Fungal Infection

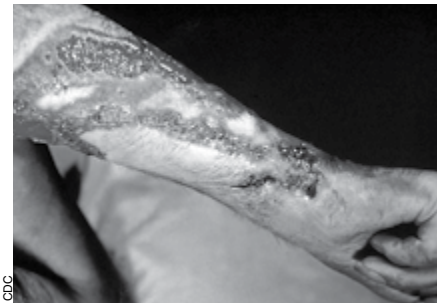


CDC

Systemic Infections are usually ones that occur deep inside the body, affecting internal organs, such as the lungs, bones, heart, and urinary tract. They often start in the lungs by inhalation of the spores and spread throughout the body. e.g. candidiasis in the kidney above.



Cutaneous (superficial) infection: Infection that affects the skin, hair, nails, genital organs, and inside of mouth. Contracted through contact with spores, e.g. trichosporosis infection of the toenails above. They are slow growing and difficult to treat.



CDC

Subcutaneous Infection: Rare infection of the fatty connective tissue beneath the skin. Contracted through direct implantation of the spores into the skin via a scratch or puncture wound, e.g. sporotrichosis (above) caused by the fungus *Sporothrix schenckii*.

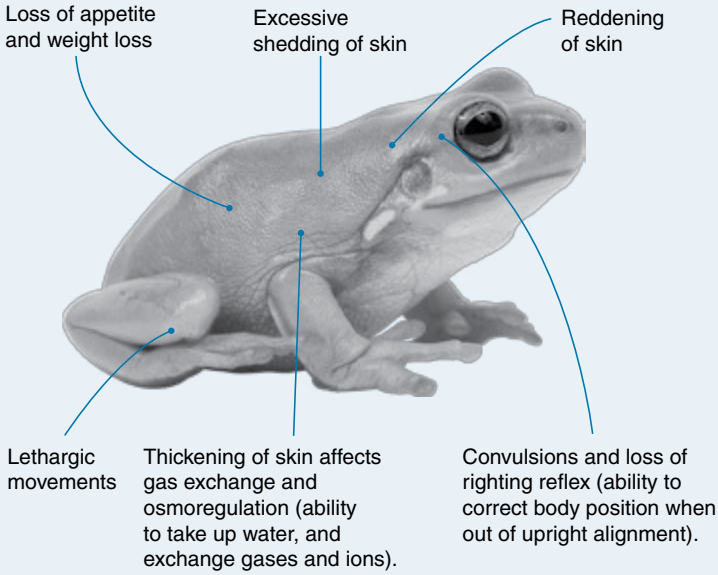
Chytridiomycosis

Chytridiomycosis is a waterborne disease of amphibians caused by the fungi *Batrachochytrium dendrobatidis*. It disperses via motile spores called zoospores, which enter the host via the skin, although much of how new hosts are infected is still unknown. It can be fatal to infected frogs within 10-18 days.

Batrachochytrium dendrobatidis is found to various parts of Australia, notably on the east coast, Adelaide, south-west Western Australia and the Kimberley region of WA.

Chytridiomycosis has been implicated in the dramatic population declines of frog species including the *Litoria nannotis* (waterfall frog), *Litoria rheocola* (common mistfrog), *Litoria spenceri* (spotted tree frog) and *Nycitmystes dayi* (lace-eyed tree frog). It is also implicated in the extinction of *Rheobatrachus silus* (southern gastric-brooding frog), *Rheobatrachus vitellinus* (northern gastric-brooding frog), *Taudactylus acutirostris* (sharp-snouted day frog), and *Taudactylus diurnus* (southern day frog).

Symptoms of Chytridiomycosis



1. Describe two features of fungal diseases: _____

_____

2. (a) Suggest one way in which chytridiomycosis causes death in infected frogs: _____

(b) Suggest why amphibians are so vulnerable to chytridiomycosis: _____

_____

(c) Frogs are popular aquarium pets in many countries. How might this be contributing to the spread of the disease?

_____

_____

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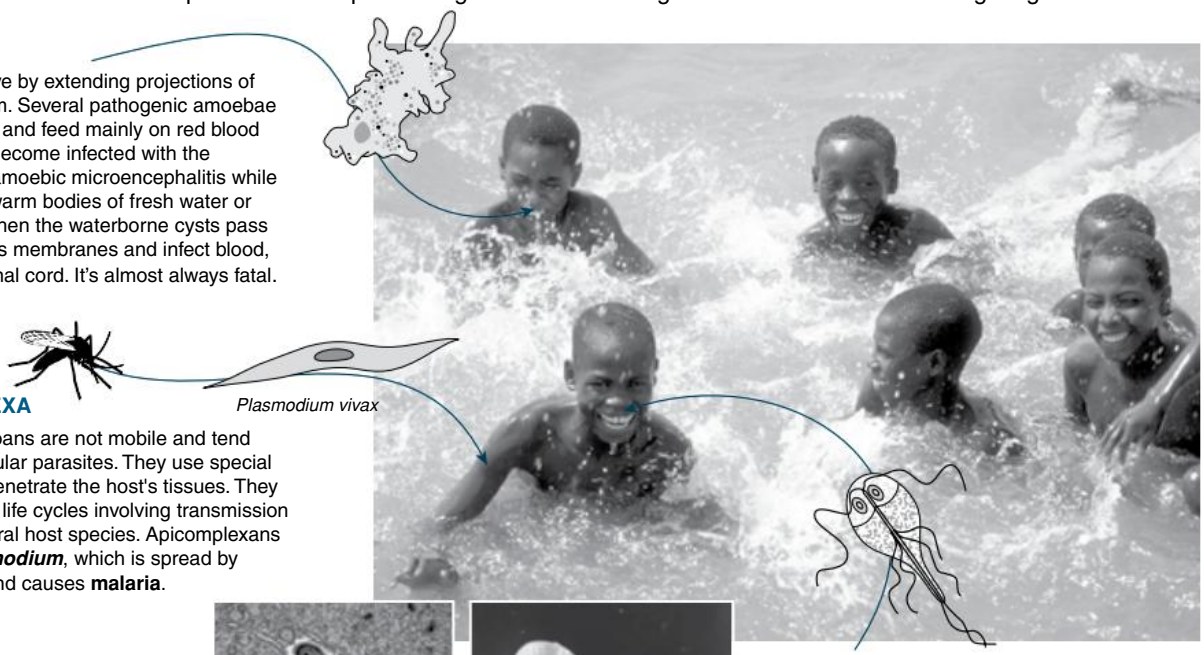
81 Protistan Diseases

Key Idea: Protistans are a large and extremely diverse group of eukaryotic organisms. A number of species are significant pathogens of animals or plants.

The protists are a group of unicellular or colonial eukaryotes. Most inhabit water or soil habitats and relatively few cause disease. However, a number of species are highly specialised pathogens. These include species of the parasitic genus *Plasmodium*, which cause malaria in humans, and species of the oomycete genus *Phytophthora*, which cause devastating dieback and blight in a number of plant species. Pathogenic protists have very complex life cycles, often involving a number of different hosts and several different life stages. Both oomycetes and plasmodia, for example, have infective motile stages as well as resistant resting stages.

AMOEBAE

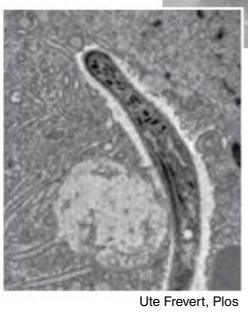
Amoebae move by extending projections of their cytoplasm. Several pathogenic amoebae infect humans and feed mainly on red blood cells. People become infected with the pathogen for amoebic microencephalitis while swimming in warm bodies of fresh water or hot springs, when the waterborne cysts pass across mucous membranes and infect blood, brain, and spinal cord. It's almost always fatal.



APICOMPLEXA

Plasmodium vivax

These protozoans are not mobile and tend to be intracellular parasites. They use special enzymes to penetrate the host's tissues. They have complex life cycles involving transmission between several host species. Apicomplexans include ***Plasmodium***, which is spread by mosquitoes and causes **malaria**.



Plasmodium sporozoite moving through the cytoplasm of the intestinal epithelia

Ute Frevert, Plos



Giardia trophozoite, SEM

CDC

FLAGELLATES

Flagellates are usually spindle-shaped, with flagella projecting from the front end. The whiplike motion of the flagella pulls the cells through their environment. *Giardia* (left) is found in the small intestine of mammals. It is passed in the faeces and its life cycle alternates between an actively swimming trophozoite (left) and an infective, resistant cyst.

Phytophthora Dieback

Phytophthora dieback is caused by the soil-borne water mould *Phytophthora cinnamomi*. Although it was originally classified as a fungus, *Phytophthora* is now included in the Protista. Flagellated zoospores enter the plant near the growing tip of the roots where they germinate, produce fungal-like hyphae, and absorb carbohydrates in the root. This eventually destroys the internal structure of the roots and causes the death of the plant.

Phytophthora cinnamomi is one of the world's most invasive plant pathogens. In Australia, it is responsible for the dieback of *Eucalyptus* trees especially in the Jarrah Forest bioregion of Western Australia. Phytophthora dieback can be treated with various fungicides including the use of phosphite salts (e.g. calcium phosphite).



Edward L. Barnard, Florida Department of Agriculture and Consumer Services, Bugwood.org

Phytophthora infection in a pine tree. Note the stunted growth. High water tables and excess irrigation provide suitable conditions root infections.



John H. Ghent, USDA Forest Service, Bugwood.org

Phytophthora infection in a pine tree showing rotted area near roots. Once a host is infected, water flow through the xylem is reduced via wilt-inducing toxins.

1. Several parasitic protozoans causing diseases in humans use other animal species as hosts for part of their life cycle. Identify the host (including class and genus) that is involved in part of the life cycle for malaria:

_____

2. Why does infection by *Phytophthora cinnamomi* cause stunted growth and death plants? _____

_____

_____

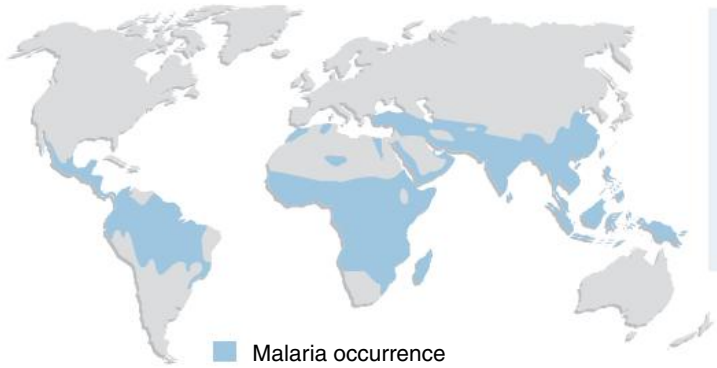
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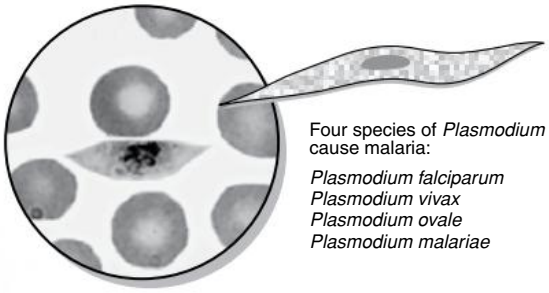
82 Malaria: A Protozoan Disease

Key Idea: Malaria is caused by parasitic protozoa. The protozoa are carried by the *Anopheles* mosquito and are passed to humans when they are bitten by the mosquito.

Malaria is a serious disease caused by **protozoan parasites** called **plasmodia**. The plasmodia have a life cycle involving two hosts, *Anopheles* mosquitoes and humans. Humans become infected when bitten by mosquitoes infected with the protozoans. In their human host, the plasmodia infect red blood cells and multiply inside the cells by asexual reproduction. Four *Plasmodium* species can cause malaria, with varying degrees of severity. Symptoms include headache, shaking, chills, fever, and even death. *Falciparum* malaria is the most severe because it affects all ages of red blood cells. Destruction of the red blood cells results in *haemolytic anaemia*. The infected blood cells also become sticky and block blood vessels to vital organs such as the kidneys and the brain.

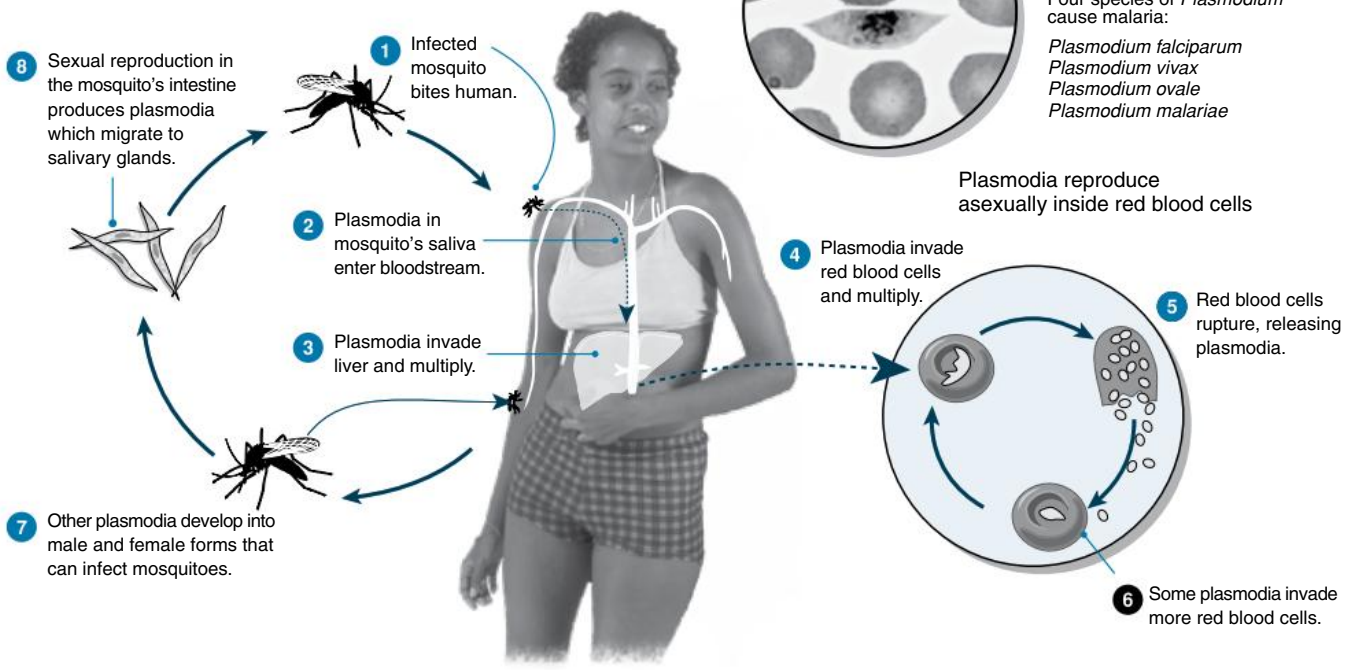


Malaria is a major health problem in tropical regions where the climate is warm and wet enough to support breeding populations of the mosquito vector. Malaria affects more than 300 million people a year in equatorial regions (left). Cases in Australia only result when infected travellers return from these regions, although Northern Australia could harbour malarial mosquitoes if the global climate warms significantly.



Four species of *Plasmodium* cause malaria:

Plasmodium falciparum
Plasmodium vivax
Plasmodium ovale
Plasmodium malariae



Plasmodia reproduce asexually inside red blood cells

1. How does a plasmodium parasite enter the body? _____

_____

2. What aspects of the biology of this pathogen could make it difficult to control? _____

_____

_____

3. (a) What biological factors are important in the global occurrence of malaria? _____

_____

(b) What measures could be cost effective in controlling the number of new malaria infections? _____

_____

4. Why is global warming expected to increase the geographical range of malaria? _____

_____

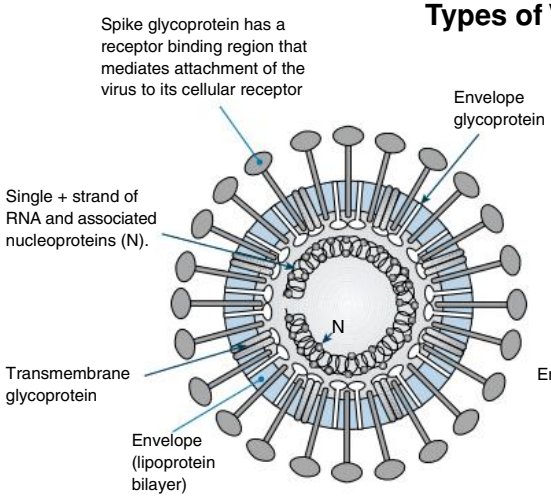
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83 Viral Diseases

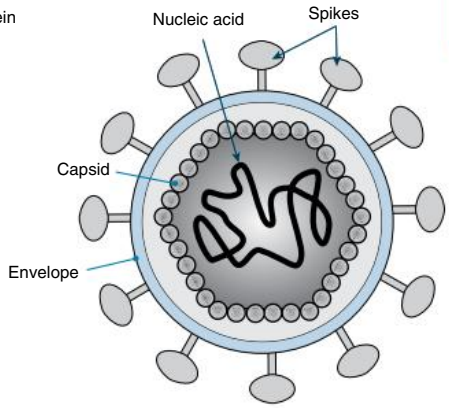
Key Idea: Viruses are infectious agents and are responsible for a large number of diseases. They infect living cells and use the cell's replication machinery to copy themselves.

A **virus** is a highly infectious pathogen that replicates only inside the living cells of other organisms. Viral infections cause a wide range of commonly occurring diseases, e.g. the common cold, influenza, cold sores, and life-threatening diseases such as HIV/AIDS and rabies. Viral infections can spread rapidly and, once contracted, are difficult to treat and must usually are left to run their course. Recovery from infection is usually associated with the host's immune system combating the infection. Developing effective antiviral drugs is difficult because they must target the virus but not affect the host cell. Antibiotics are ineffective against viruses because they only target specific aspects of bacterial metabolism. Currently, immunization to induce an immune response provides the best protection against viral disease. However, viruses mutate rapidly and this immunity may not be lifelong.

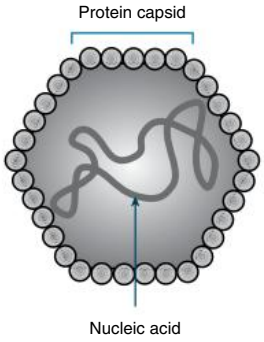
Types of Viruses Affecting Humans



Structure of a coronavirus



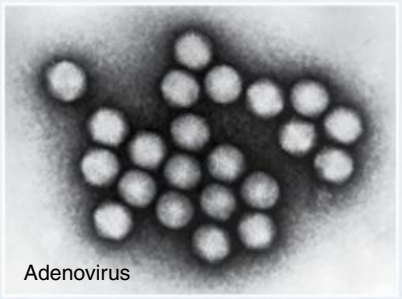
General structure of an enveloped animal virus, e.g. Herpesvirus



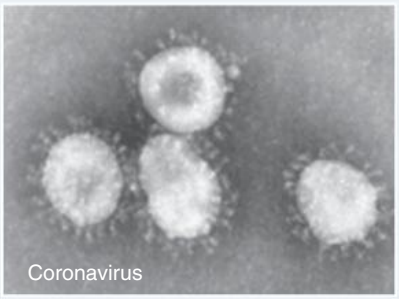
General structure of a non-enveloped animal virus, e.g. Papillomavirus (wart virus

Viruses are not organisms!

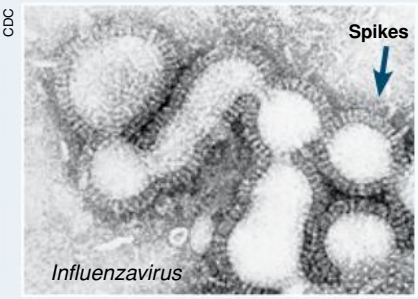
Viruses are metabolically inert until they are inside the host cell and hijacking its metabolic machinery to make new viral particles.



Adenoviruses are medium-sized (90-100 nm), non-enveloped viruses containing double-stranded DNA. They cause respiratory illness and are very stable to chemical or physical agents, allowing for prolonged survival outside of the body.



Coronaviruses infect the upper respiratory and gastrointestinal tracts of birds and mammals, including humans. Their name comes from the crown or corona of spikes and they have the largest genome of any of the single stranded RNA viruses.



Some viruses have an envelope around the capsid, which protects the virus from the host's nuclease enzymes. Spikes on the envelope provide a site for attachment to the host. ***Influenzavirus*** has many of these glycoprotein spikes.

1. Explain the basis of host specificity in viruses: _____

_____

_____

2. (a) What is the most effective method of controlling viral diseases? _____

(b) Explain your answer: _____

(c) Discuss limitations to this approach: _____

_____

_____

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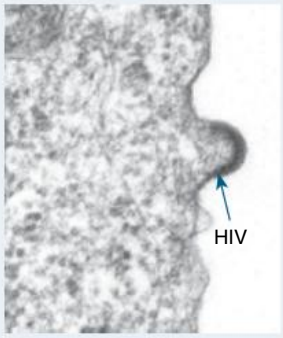
LINK 89 LINK 84 LINK 74 WEB 83 KNOW

84 HIV and AIDS

Key Idea: The human immunodeficiency virus infects lymphocyte cells, eventually causing AIDS, a fatal disease, which acts by impairing the immune system.

HIV (human immunodeficiency virus) is a retrovirus, a single-stranded RNA virus which infects lymphocytes called helper T-cells. Over time, a disease called **AIDS** (acquired immunodeficiency syndrome) develops and the immune system loses its ability to fight off infections as more helper T-cells are destroyed. There is no cure or vaccine for HIV, but some drugs have been developed which can slow the progress of the disease.

HIV Infects Lymphocytes



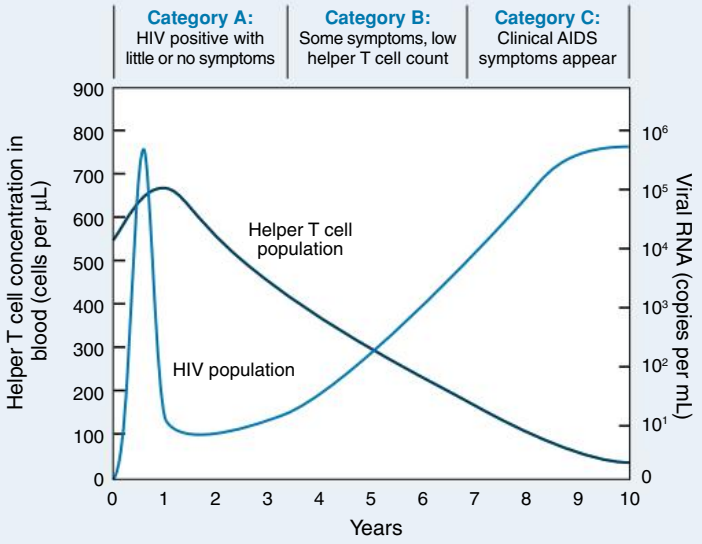
CDC

HIV budding from a lymphocyte

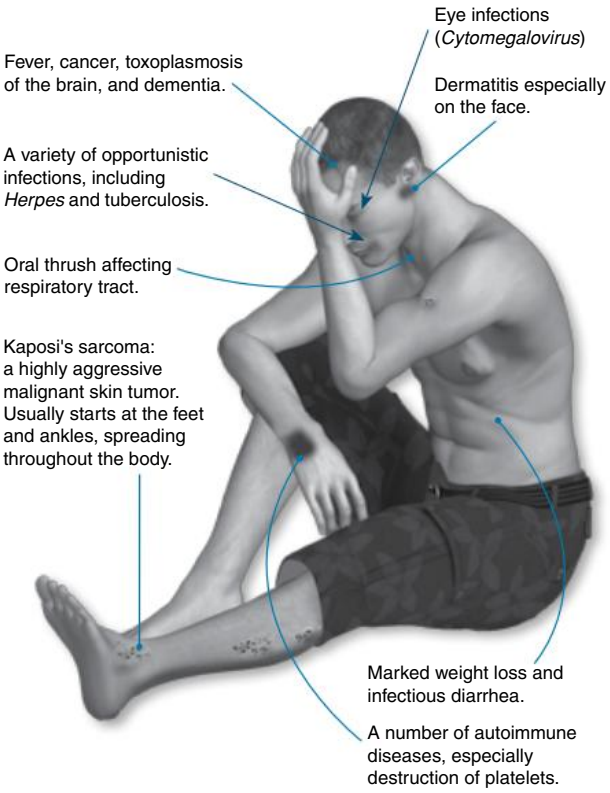
HIV infects helper T-cell lymphocytes. It uses the cells to replicate itself in great numbers, then the newly formed viral particles exit the cell to infect more helper T-cells. Many helper T-cells are destroyed in the process of HIV replication.

Helper T-cells are part of the body's immune system, so when their levels become too low, the immune system can no longer fight off infections.

The graph below shows the relationship between the level of HIV infection and the number of helper T-cells in an individual.



AIDS: The End Stage of an HIV Infection



The range of symptoms resulting from HIV infection is huge, but are not the result of the HIV infection directly. The symptoms arise from secondary infections that gain a foothold in the body due to the weakened immune system (due to the reduced number of helper T-cells). People with healthy immune systems can be exposed to pathogens and not suffer serious effects because their immune system fights them off. However, people with HIV are susceptible to all pathogens because their immune system is too weak to fight them off. As the immune system become progressively weaker, the infected person becomes sicker.

1. Consult the graph above showing the stages of HIV infection (remember, HIV infects and destroys helper T cells).

   (a) How do viral numbers change with the progression of the disease? _____

   _____

   _____

   (b) How do the helper T cells respond to the infection? _____

   _____

   _____

2. Why does the HIV virus have such a devastating effect on the human body's ability to fight disease?

   _____

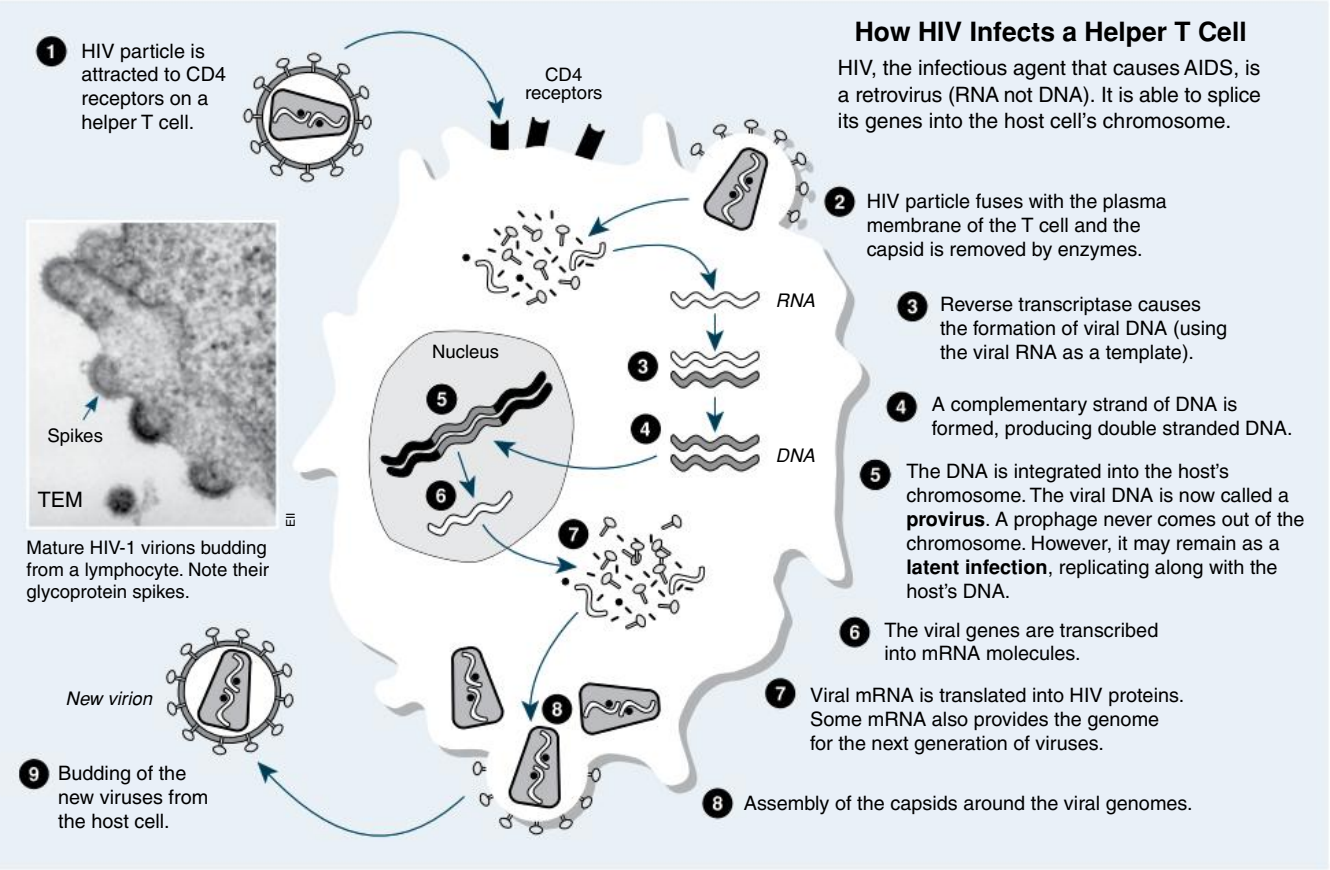
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85 Replication in HIV

Key Idea: HIV uses reverse transcriptase to synthesise DNA which is then integrated with the host cell's DNA.

Animal viruses exhibit a number of different mechanisms for **replicating**, i.e. entering a host cell and producing and releasing new virions. Enveloped viruses bud out from the host cell, whereas those without an envelope are released by rupture of the cell membrane. Three processes (attachment, penetration, and uncoating) are shared by both DNA- and RNA containing animal viruses. The example below describes replication in the retrovirus HIV, where the virus uses its own reverse transcriptase to synthesise viral DNA and produce **latent proviruses** or active, mature retroviruses.



1. (a) How does an HIV particle enter a host cell? _____

   _____

   (b) What is the role of reverse transcriptase in the life cycle of a retrovirus? _____

   _____

   _____

   (c) Explain the significance of the formation of a provirus: _____

   _____

   _____

2. Summarise the steps involved in invasion of a host cell by an enveloped viral particle such as *Influenzavirus*:

   (a) Attachment: _____

   (b) Penetration: _____

   (c) Uncoating: _____

   (d) Biosynthesis: _____

   (e) Release: _____

3. Some of the drugs for treating HIV inhibit reverse transcriptase. Why is this an effective strategy? _____

   _____

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86 Human Viral Diseases

Key Idea: There is a wide range of human diseases caused by viruses, including influenza and Ross River fever.

Viruses are responsible for a number of human diseases including polio, hepatitis, measles, and HIV/AIDS. Most common viral diseases in humans spread easily between hosts (they are contagious). Although some viral diseases can be controlled effectively by vaccination, this is not always permanently effective because viruses undergo frequent genetic changes and new strains may appear seasonally (e.g. influenza). While many viruses are contagious, others are transmitted by a vector, e.g. Ross River virus is carried by mosquitoes. A vector-borne disease occurs in regions where the environment supports the proliferation of the vector, e.g. mosquitoes breed in warm, wet environments.

Ross River Virus

Ross River virus (RRV) is endemic to Australia, Papua New Guinea, and several other islands of the South Pacific. It is a single strand RNA alphavirus and causes Ross River fever. RRV is transmitted by mosquitoes and may have natural reservoirs in native Australian mammals. In Australia, most RRV infections occur in the wetter tropical areas of QLD, WA, and NT. There have been several large outbreaks since the first noted outbreak in 1928. The largest outbreak occurred in 1979-80 across the Western Pacific, affecting 60,000 people.

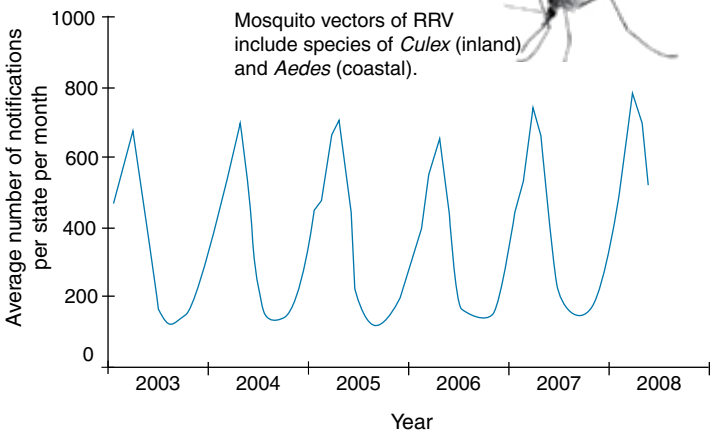
Ross River Fever

Symptoms of Ross River fever include fever, rash, and arthritic-like symptoms causing extreme pain in various joints. The disease has a greater impact in adults than children, who may not show any symptoms. In most cases, the disease lasts about a month but can reoccur over a period of years. Currently a blood test is the only way to confirm Ross River fever and there is no vaccine. Cases are more prevalent from January to March during the summer/autumn wet season.

Notifications of Ross River Virus in Australia, 2003 - 2008



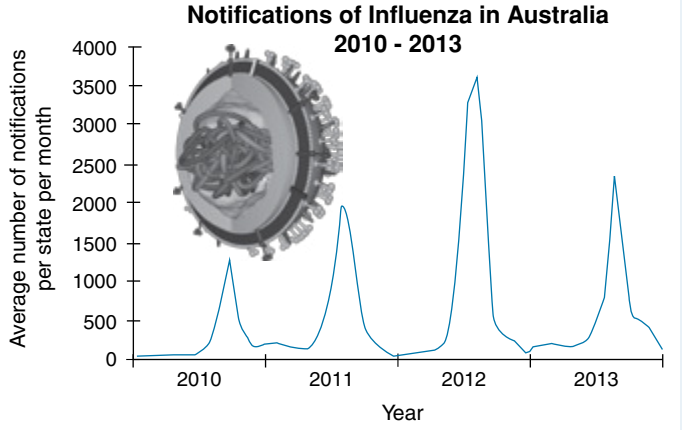
Mosquito vectors of RRV include species of *Culex* (inland) and *Aedes* (coastal).



Influenza

Although it is often looked upon as simply a bad cold, influenza (commonly called "the flu") has arguably caused more human deaths than any other disease throughout history. Influenza occurs in seasonal epidemics, usually infecting around five million people globally, and causing the death of between 250,000 and 500,000 people annually. The influenza virus continually changes in two ways. **Antigenic drift** involves small cumulative genetic changes over time. Occasionally the virus will also undergo **antigenic shift** producing an entirely new strain that causes a pandemic. The latest of these is the H1N1/09 strain, commonly referred to as swine flu. This strain caused around 14,000 deaths globally. Compare this the Spanish flu pandemic of 1918-1920, during which possibly up to 100 million people died globally (more than all deaths during World War I and possibly as many as World War II).

Notifications of Influenza in Australia 2010 - 2013



1. (a) How is the Ross River virus transmitted to people? _____

   (b) Suggest why cases of Ross River fever are more prevalent in January to March? _____

   _____

   _____

   (c) There was a large mosquito outbreak in 2013 in Newcastle NSW. Predict the effect of this on the prevalence of Ross River fever:

   _____

2. (a) What time of the year is the influenza virus most prevalent? _____

   (b) Why must people receive the flu vaccine every year if they want to remain protected against influenza?

   _____

   _____

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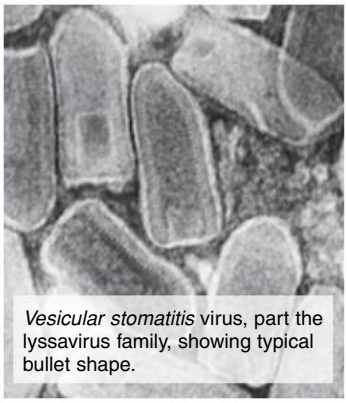
87 Animal Viral Diseases

Key Idea: Animals can be reservoirs of disease that can spread to humans. Viruses can have a detrimental economic impact if commercially important species are affected.

Many pathogens can be transmitted from animals to people (**zoonosis**). Many of these are endemic to certain animal populations and only occasionally are transmitted to humans through direct contact such as a scratch or a bite. A number of deadly viruses are transmitted to humans in this way. The Ebola virus resides in populations of fruit bats or monkeys in Africa. In humans, it causes severe haemorrhaging and death. In Australia, the Australian bat lyssavirus is present in a number of native mammals including native bats. Humans can be affected by viruses economically. Viruses of honey bees can kill the hive and reduce production and pollination of crops.

Australian Bat Lyssavirus

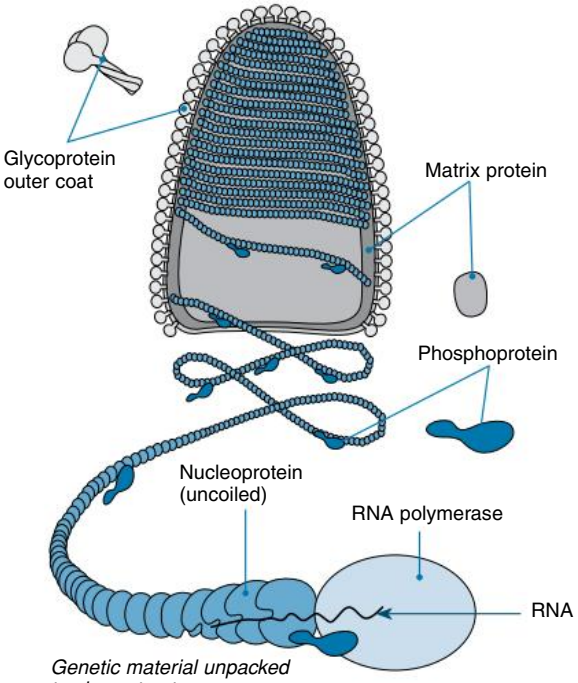
Australian bat lyssavirus (ABL) (right) was first identified in a juvenile black flying fox in 1995. It is closely related to the rabies virus (not found in Australia) and can be transferred to humans if they are bitten or scratched by an infected bat. Like the rabies virus, ABL affects the central nervous system. The virus enters peripheral nerves and replicates, spreading up the nerves to the brain, at which point symptoms (convulsions, possible aggression, and paralysis) begin to appear. Death may occur shortly after the symptoms appear. The virus also affects nerves innervating the salivary glands, causing them to produce large amounts of saliva. A person bitten by an infected bat may not show symptoms for some time (in one case over two years). However, once symptoms do appear mortality is currently 100%. Of the three cases of human infection noted in Australia, all have been fatal.



Vesicular stomatitis virus, part the lyssavirus family, showing typical bullet shape.

CDC

ABL has the typical bullet shape of all lyssaviruses. The genetic material is stored as RNA and includes coding for five proteins (nucleoprotein (N), phosphoprotein (P), matrix protein (M), glycoprotein (G) and polymerase (L)). The RNA is transcribed into mRNA by the polymerase enzyme bound in the virus.



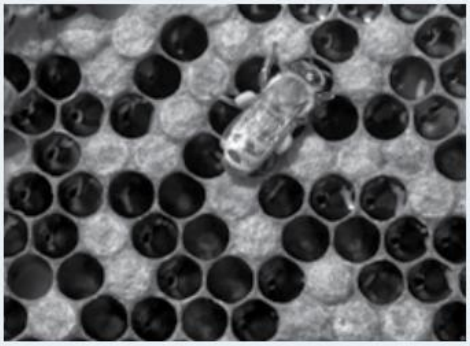
Genetic material unpacked to show structure

Honey Bee Viruses

There are around 24 known honey bee viruses. In many cases they are asymptomatic (produce no symptoms) when the infection is at low levels. However, under certain circumstances the health of the hive can be severely affected. The most important and common honey bee viruses include sacbrood, black queen cell virus, Kashmir bee virus, and paralysis virus.

Sacbrood is one of the most widely distributed honey bee viruses. Infected larvae die just before pupation. The dead larvae turn light to dark brown and are surrounded by a liquid filled bag containing millions of virus particles. Removal of the dead larvae by workers infects more workers and spreads the disease through the colony. This can cause the loss of the colony resulting in lower pollination rates and poor crop production.

There are no known cures for any of the bee viruses. Management of viruses is aimed at prevention of infection by careful management of hives including regulation of the honey combs and choosing queens from resistant stock.



Comb of a healthy hive. In a hive with sacbrood, partially uncapped cells are scattered among healthy brood cells.

1. Which part of the body does ABL mainly affect? _____

2. What is the mortality rate for ABL infections? _____

3. A person infected by the Australian bat lyssavirus can show aggression and produce a lot of saliva. What is the benefit of this to the virus?

_____

_____

4. Why is the loss of bee hives through disease of economic importance? _____

_____

_____

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88 Prions

Key Idea: Prions are misfolded infectious proteins that can propagate by causing misfolding in the original protein type.

Until recently, all pathogens were thought to contain some form of nucleic acid. We now know that particular proteins, called **prions**, are capable of causing infection. Prions have been spread by eating contaminated meat and, because they resist normal sterilisation methods, they can be spread on surgical instruments. Prions are produced by mutations in the gene coding for a normal cell protein (PrP). They cause a group of degenerative nervous diseases in mammals called transmissible spongiform encephalopathies (TSE). They include scrapie in sheep, **BSE** in cattle, and **kuru** in humans. Different mutations of the PrP gene are responsible in each case.



The disease **kuru** first brought prion diseases to prominence in the 1950s. It occurred in the geographically isolated tribes in the Fore highlands of Papua New Guinea. These people were eating the brain tissue of dead relatives for religious reasons.

Normal and infectious prions have the same primary structure, so go unchallenged by the immune system. If the infectious prion is from another species (as in vCJD) there is an initial immune response but this is shut down as the infectious protein converts more and more of the body's own PrP^C to PrP^{SC}.

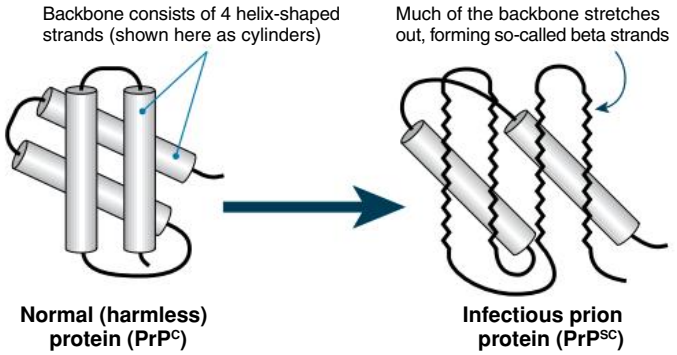
Prion diseases of humans (and cause)

All these diseases are characterised by dementia and loss of coordination. There may be other symptoms as well.

- Kuru (infection through cannibalism)
- Variant Creutzfeldt-Jacob Disease (vCJD) (infection)
- Classical Creutzfeldt-Jacob Disease (infection or mutation)
- Fatal Familial Insomnia (inherited mutation)

Infectious Prion Proteins

A shape change transforms the harmless protein into an infectious **prion**. The change may be caused by a point mutation in the encoding gene. The normal (common) form of the protein is denoted PrP^C, whereas the abnormal form is denoted PrP^{SC} (after scrapie, the prototype prion).

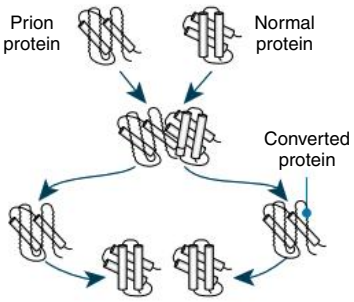


Propagation of the Prion Protein

Infectious protein (prion) has an unusual shape and binds to the normal form of the protein.

The normal protein is converted to the infectious prion.

The original and newly formed prions attack other nearby proteins. In time, prions accumulate to dangerous levels.



1. What is the main feature of prions distinguishing them from other infectious agents? _____

2. How does a prion's mode of transmission make it a successful agent of disease? _____

3. What is the source of infection for people with **variant CJD**? _____

4. How did the cultural practices of highland tribes in PNG enable the spread of **kuru**? _____

5. An epidemic of BSE in the UK in the 1990s had its origin in the practice of processing waste parts of cattle (particularly nervous tissue) and recycling them into cattle feed. Infected cattle subsequently entered the human food chain and were linked to cases of vCJD. Explain why it is poor practice to process an animal and feed it back to the same species:

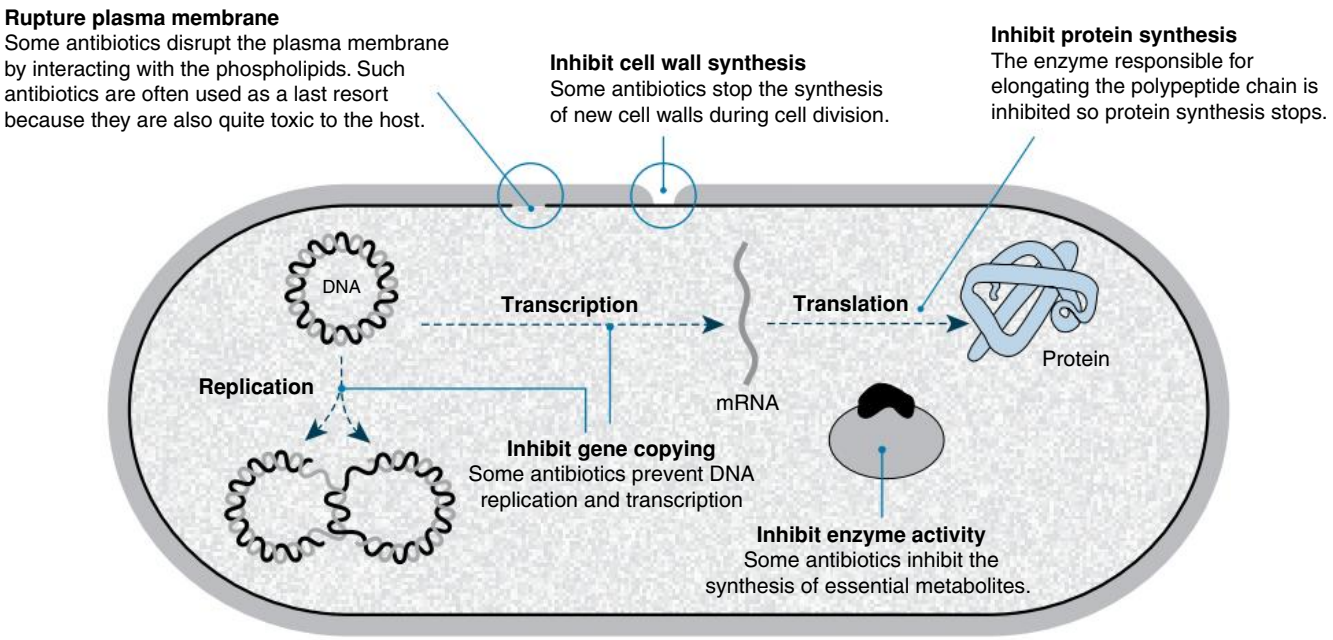
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89 Antimicrobial Drugs

Key Idea: Antibiotics work against bacteria but are ineffective against viral pathogens, which require special antiviral drugs.

An **antibiotic** is a substance that inhibits or prevents bacterial growth. Antibiotics are produced naturally by bacteria and fungi against their competitors, and this ability has been exploited by humans for the control of bacterial infections. Antibiotics interfere with bacterial growth by interfering with key aspects of bacterial metabolism (below). Their action may kill the bacterial cells directly (**bactericidal**) or simply inhibit their growth (**bacteriostatic**). The ideal antimicrobial drug has **selective toxicity**, killing the pathogen without damaging the host. The best antibiotics have a narrow spectrum of activity and target only the bacterial species responsible for the infection. Broad spectrum antibiotics are active against a large number of bacterial species and so affect the host's natural microflora as well as the pathogen. While antibiotics are effective against bacteria, they are not effective against viruses. Antiviral drugs are used to inhibit various stages of the viral life cycle, from penetrating the host cell to disrupting the release of the virus.

How Antibiotics Work



1. (a) What are the requirements of an "ideal" antimicrobial drug? _____

   _____

   _____

   (b) In what way do antibiotics satisfy these requirements? _____

   _____

   _____

2. Why are antibiotics that target the plasma membrane also harmful to the host? _____

   _____

   _____

3. Explain the difference between bacteriostatic and bactericidal antibiotics: _____

   _____

   _____

   _____

4. Explain why antibiotics do not work on viruses: _____

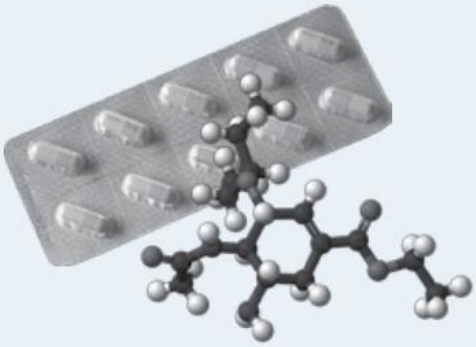
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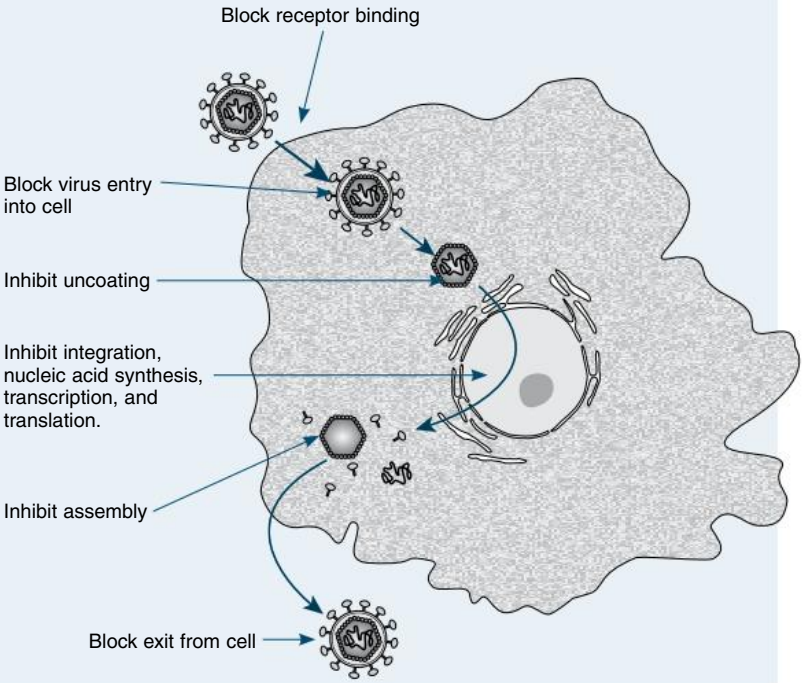
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Mechanisms of Antiviral Drugs

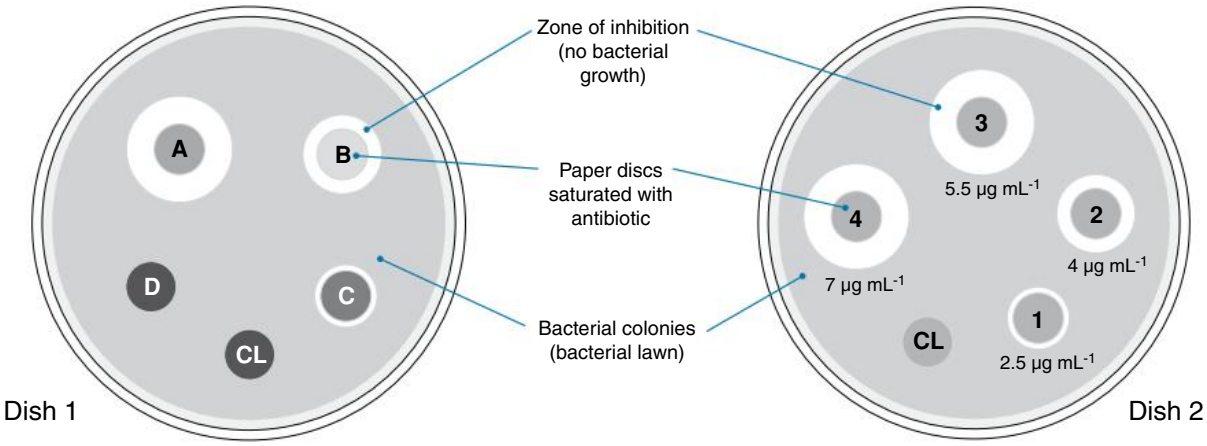
Currently, antiviral drugs are available for use against HIV, hepatitis, influenza, and the herpes virus. Most antiviral drugs work by inhibiting replication of the virus. The drug stops the virus replicating giving the immune system time to destroy the virus. Antiviral drugs work on several parts of virus's life cycle (right).



Oseltamivir (marketed as Tamiflu, above) slows the spread of influenza A and B viruses in the body by preventing a newly formed virus particle from budding from an infected cell. However, various trials have shed doubt of whether the drug produces any real benefit to flu sufferers.



5. Two students investigated the effect of antibiotics on bacterial growth. They placed antibiotic discs on petri dishes evenly coated with bacterial colonies. Dish 1 contained four different antibiotics (A-D) and a control (CL). Dish 2 contained four different concentrations of a single antibiotic and a control (CL).



(a) Which was the most effective antibiotic on Dish 1? Explain your choice: _____

(b) Which was the most effective concentration on Dish 2? Explain your choice: _____

_____

_____

6. For each of the following mechanisms used by antiviral drugs explain how it prevents the virus's spread:

(a) Blocking receptor binding: _____

_____

_____

(b) Inhibiting assembly: _____

_____

_____

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90 Containing the Spread of Disease

Key Idea: Preventing the entry and spread of pathogens is important in protecting a country's population and industries from infectious diseases.

Many factors can influence the spread of disease, including the social climate, diet, general health, and access to medical care. Human intervention and modification of behaviour, including vaccination, can reduce the transmission rate of some diseases and inhibit their spread. Global air travel and international trade in commodities has increased the risk that diseases of humans, livestock, and crops will be spread between countries. Australia is fortunate in that its geographical isolation has helped to prevent the spread of disease from other parts of the world. Precautions such as quarantine, which isolates exposed individuals that may be infected, as well as screening of imported produce and international travellers, help to limit the entry of diseases into Australia. Quarantine is distinct from isolation, which aims to contain disease by isolating an already infected person.



Transmission of disease can be reduced by adopting 'safe' behaviours. Examples include using condoms to reduce the spread of STIs, isolating people already infected, or establishing quarantine procedures for people who have been exposed to infection.



CDC

The environment can be made less suitable for the growth and transmission of pathogens. For example, spraying drainage ditches and draining swamps eliminates breeding habitats for mosquitoes carrying diseases such as malaria and dengue fever.



Disinfectants and sterilisation techniques, such as autoclaving, destroy pathogenic microbes before they have the opportunity to infect. The use of these techniques in medicine has significantly reduced post operative infections and associated deaths.

The Role of Quarantine

The **Australian Quarantine Inspection Service** (AQIS) plays an important role preventing the entry of exotic pests and diseases that could affect plant, animal, and human health, and the environment. All passengers and freight entering Australia by air or sea (even small, private vessels) are required to pass inspection by a Quarantine Officer. Australia has strict quarantine regulations because introduced animals and plants could also bring in pests and diseases that have no natural enemies or controls.

Goods PROHIBITED entry into Australia without authorisation

- Milk and dairy products
- Seeds and beans
- Popping corn and raw nuts
- Eggs and egg products
- Fresh fruit and vegetables
- Live animals
- Meat and all pork products
- Salmon and trout products
- Live plants
- Biological materials (e.g. human/animal vaccines)
- Deer horn/velvet
- Soil and sand



The Disease Risk

Communicable diseases spread from one host to another, either directly or indirectly. Those that are easily spread, such as measles, are said to be contagious. Such diseases are a threat to public health and many must be notified to health authorities. Many of the serious contagious bacterial and viral diseases of the past are now controlled through vaccination programmes. Even if susceptible individuals contract the disease, the spread is limited because of the herd immunity of the generally vaccinated population.

Noncommunicable diseases, such as chronic lung diseases, are not spread from one host to another and pose less threat to public health. A disease that occurs only occasionally and is restricted in its spread is called a sporadic disease (e.g. Legionnaire's disease).





English measles (top) and whooping cough are both controlled by vaccination

1. (a) Explain why contagious diseases present such a threat to public health:_____

(b) How are contagious diseases generally best controlled?_____

(c) Why is this method of control effective in limiting the spread of the disease?_____

2. How does quarantine prevent the spread of disease? _____

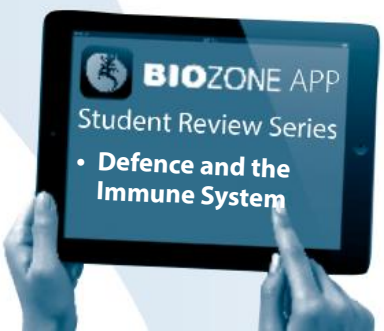
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QLD As required
VCE Unit 3 (2)
WA ATAR Unit 4
SA Theme O

Defence and the Immune System

Key terms

active immunity
allergic reaction
antibody (=immunoglobulin)
antigen
asthma
autoimmune disease
B cell (=B lymphocyte)
cell-mediated immunity
clonal selection
fever
hayfever
histamine
humoral immunity
hypersensitivity
immunity
immunological memory
infection
inflammation
interferon
leucocyte
lymphocyte
macrophage
MHC
monoclonal antibody
non-specific defences (=innate immunity)
passive immunity
phagocyte
primary response
secondary response
specific (=adaptive) immune response
T cell (=T lymphocyte)
thymus
vaccination (=immunisation)

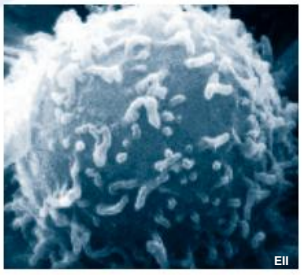


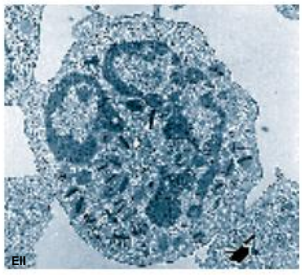
The body's layers of defence

Learning aims and skills

Activity number

- ☐ 1 Describe the range of physical and chemical defences in animals. — 91
- ☐ 2 Explain how the body distinguishes self from non-self. Include reference to the role of the major histocompatibility complex (MHC) and its role in self-recognition and in determining tissue compatibility in transplant recipients. — 92
- ☐ 3 Explain the basis of the Rh and ABO blood group systems in humans. Explain the consequences of blood type incompatibility in blood transfusions. — 93
- ☐ 4 Describe non-specific (innate) defences in humans, describing the nature and role of each of the following in protecting against pathogens: — 94 -97
  (a) Skin (including sweat and sebum production) and mucous membranes.
  (b) Body secretions (tears, urine, saliva, gastric juice).
  (c) Natural anti-bacterial and anti-viral proteins, e.g. interferon.
  (d) Phagocytosis, the inflammatory response, fever, and cell death.





Specific immunity

Learning aims and skills

Activity number

- ☐ 5 Describe the specific immune response, including the role of specificity and memory. Describe cell-mediated immunity and humoral (antibody-mediated) immunity, identifying the specific white blood cells involved in each case. — 98 99
- ☐ 6 Describe clonal selection and the basis of immunological memory. Explain how the immune system is able to respond to the large range of potential antigens. — 100
- ☐ 7 Explain antibody production, including how B cells bring about humoral (antibody-mediated) immunity to specific antigens — 101
- ☐ 8 Distinguish between naturally acquired and artificially acquired immunity and between active and passive immunity. — 102
- ☐ 9 Explain the principles of vaccination, including reference to the primary and secondary response to infection and the role of these. — 103
- ☐ 10 Describe the production and applications of monoclonal antibodies. — 104 105

Immune dysfunction and disease

Learning aims and skills

Activity number

- ☐ 11 With reference to specific examples, describe the result of immune system dysfunction, e.g. in hypersensitivity reactions such as asthma and hayfever, and in autoimmune diseases such as multiple sclerosis. — 106 107

Defence mechanisms in plants

Learning aims and skills

Activity number

- ☐ 12 Describe how plants inhibit disease and recover from infection. Describe passive and active defences in plants, including reference to physical and chemical barriers, cellular defences, and cell death (necrosis). — 108

91 Chemical Defences In Animals

Key Idea: Animals have evolved a wide range of chemical defences that protect against infection by pathogens.

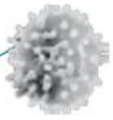
Living organisms are under constant attack from many different **pathogens** (disease causing organisms). As a result, they have evolved many ways to limit infection by pathogens. Plants and animals have a number of defences, including **chemical defences**, to destroy pathogens. Invertebrates were initially thought to lack specific immunity, but recent findings suggest the opposite is true, although it is different to that seen in vertebrates.

Vertebrate Defences

Antimicrobial substances: Chemicals (e.g. **lactoferrin**) are secreted from the skin and other body fluids, and also by some white blood cells. These kill pathogens or inhibit their growth.

Lysozyme in tears, saliva, milk, and mucus.

Antimicrobial peptides: **Defensins** are abundant in phagocytes and small intestinal mucosa of mammals.



Specific immune response: Lymphocytes recognise and destroy specific pathogens.

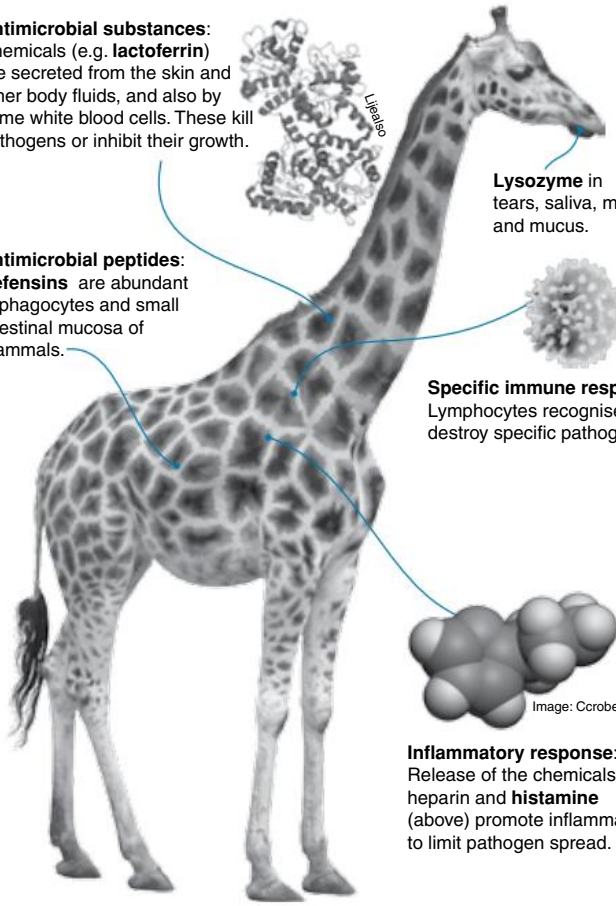


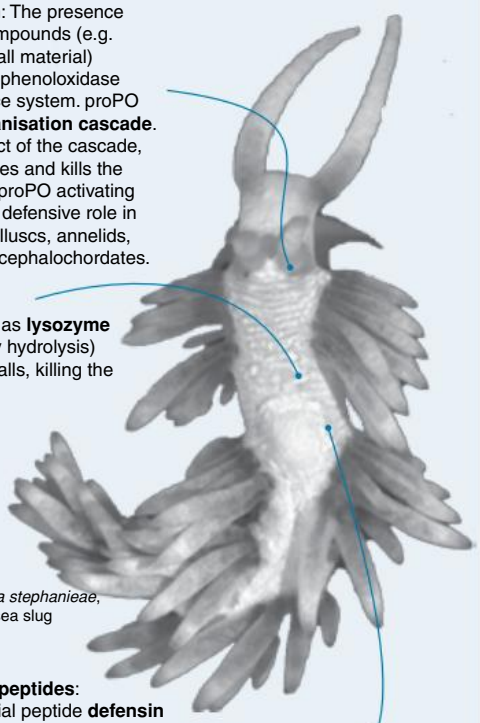
Image: Ccroberts

Inflammatory response: Release of the chemicals heparin and **histamine** (above) promote inflammation to limit pathogen spread.

Invertebrate Defences

ProPO system: The presence of microbial compounds (e.g. bacterial cell wall material) initiates the prophenoloxidase (proPO) defence system. proPO induces a **melanisation cascade**. The end-product of the cascade, melanin, encases and kills the pathogen. The proPO activating system plays a defensive role in arthropods, molluscs, annelids, ascidians, and cephalochordates.

Lytic system: Enzymes such as **lysozyme** break down (by hydrolysis) bacterial cell walls, killing the bacterial cells.



Aeolidiella stephanieae, a sea slug

Antimicrobial peptides: The antimicrobial peptide **defensin** binds to the cell membrane of pathogens, and causes damage by puncturing the membrane. The pathogen becomes "leaky" and dies. Defensins are also found in vertebrates.

PHOTO: Kristof A. & Klussmann-Kolb A. cc 2.0



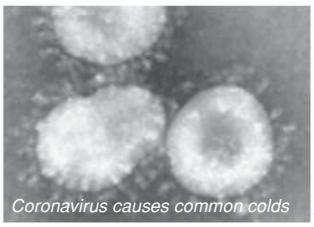
E. coli can contaminate water

Pathogenic **bacteria** cause a wide range of diseases. Some types of *E. coli* are harmless gut residents, but others can cause disease.



Mites attached to harvestman host

Vertebrates and invertebrates are host to a range of arthropod vectors, which may carry pathogenic microorganisms on their mouthparts.



Coronavirus causes common colds

Viruses are species-specific intracellular parasites and cause many common diseases. They infect all other organisms, including bacteria.



Malaria sporozoite

Eukaryotic pathogens: protozoa (e.g. the malaria parasite above), algae, fungi, and parasitic worms, may be pathogenic.

1. (a) Describe the advantage of having multiple (non-specific) defence responses: _____

_____

_____

(b) Describe a disadvantage of having only general (non-specific) defence responses: _____

_____

2. Compare and contrast the non-specific defences of vertebrate and invertebrate animals: _____

_____

_____

_____

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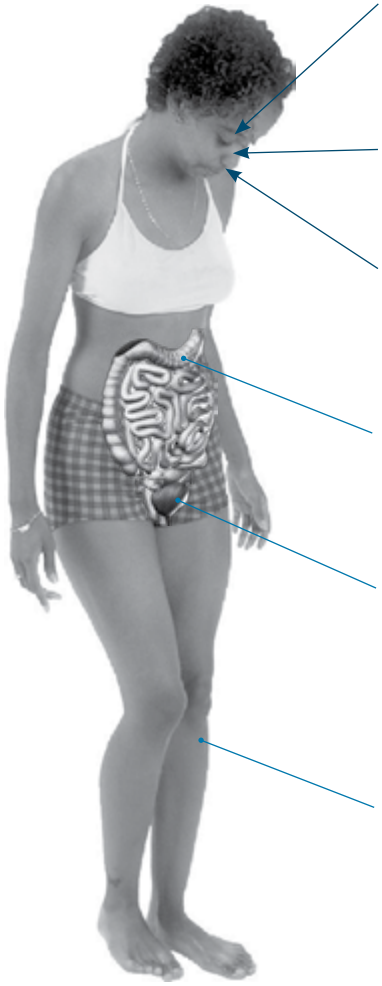
92 Targets for Defence

Key Idea: Cell surface MHC antigens enable the body to distinguish its own tissues from foreign material.

In order for the body to effectively defend against pathogens, it must first be able to recognise its own tissues (self) and distinguish itself from foreign tissue (e.g. bacteria). Failure of self/non-self recognition can lead to autoimmune disorders, in which the immune system mistakenly destroys its own tissues. The ability of the body to recognise its own molecules has implications for medical techniques such as tissue grafts, organ transplants, and blood transfusions. Foreign tissues are attacked by the body's immune system (rejected). Even a healthy pregnancy involves suppression of specific features of the self recognition system, allowing the mother to tolerate a nine month relationship with a foreign body (a fetus).

The Body's Natural Microbiota

After birth, normal and characteristic microbial populations begin to establish themselves on and in the body. A typical human body contains 1 X 10^{13} body cells and 1 X 10^{14} bacterial cells. These microbes establish more or less permanent residence but, under normal conditions, do not cause disease. In fact, this normal microflora can benefit the host by preventing the overgrowth of harmful pathogens.



Eyes: The conjuctiva, a continuation of the skin or mucous membrane, contains a similar microbiota to the skin.

Nose and throat: Harbours a variety of microbes, including streptococci and staphylcocci

Mouth: Supports a large and diverse microbiota. It is an ideal microbial environment; high in moisture, warmth, and nutrient availability.

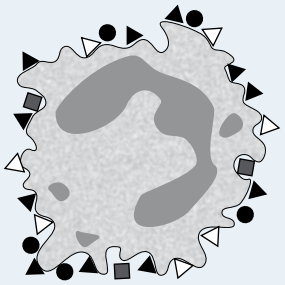
Large intestine: Contains the body's largest resident population of microbes because of its available moisture and nutrients.

Urinary and genital systems: The lower urethra in both sexes has a resident microbial population. The vagina has a population of acid-tolerant lactic acid bacteria because of the low pH nature of its secretions.

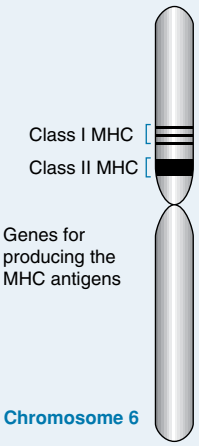
Skin: Skin secretions prevent most of the microbes on the skin from becoming residents.

Distinguishing Self from Non-Self

The human immune system achieves self-recognition through the major histocompatibility complex (**MHC**). This is a cluster of tightly linked genes on chromosome 6. These genes code for protein molecules (MHC antigens) that are attached to the surface of body cells. They are used by the immune system to recognise its own or foreign material. Class I MHC antigens are found on the surfaces of almost all human cells. Class II MHC antigens occur only on macrophages and B-cells of the immune system.



MHC surface proteins (antigens) provide a chemical signature that allows the immune system to recognize the body's own cells



Tissue Transplants

The MHC is responsible for the rejection of tissue grafts and organ transplants. Foreign MHC molecules are antigenic, causing the immune system to respond in the following way:

- T cells directly lyse the foreign cells
- Macrophages are activated by T cells and engulf foreign cells
- Antibodies are released that attack the foreign cell
- The complement system injures blood vessels supplying the graft or transplanted organ

To minimise this rejection, attempts are made to match the MHC of the organ donor to that of the recipient as closely as possible.

1. Why is it healthy to have a natural population of microbes both on and inside the body? _____

_____

2. (a) What is the nature and purpose of the **major histocompatibility complex** (MHC)? _____

_____

_____

_____

(b) Explain the importance of such a self-recognition system: _____

_____

3. Name two situations when the body's recognition of 'self' is undesirable: _____

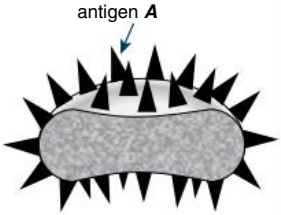
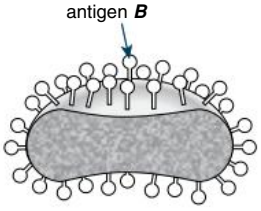
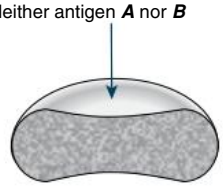
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93 Blood Group Antigens

Key Idea: The ABO classification of human blood is based on the presence or absence of inherited antigens on the surface of red blood cells.

Blood is classified into groups according to antigens on the surface of red blood cells (RBCs).**Blood typing** is essential for safe **blood transfusion**. Transfusion with incompatible blood types may cause a fatal transfusion reaction in which RBCs from the donated blood react with pre-formed antibodies in the recipient's blood. The donor RBCs clump together (agglutinate), block capillaries, and rupture (**hemolysis**). Although human RBCs have more than 500 known antigens, fewer than 30 are regularly tested for and they are not common enough to be used for cross-matching. The **ABO** and **rhesus** (Rh) blood group antigens are the best known. Rh^+ or Rh^- denotes presence or absence of the Rh antigen.

	Blood type A	Blood type B	Blood type AB	Blood type O
Antigens present on the ***red blood cells***	antigen ***A*** 	antigen ***B*** 	antigens ***A*** and ***B*** 	Neither antigen ***A*** nor ***B*** 
Anti-bodies present in the ***plasma***	 Contains **anti-B** antibodies; but no antibodies that would attack its own antigen ***A***	 Contains **anti-A** antibodies; but no antibodies that would attack its own antigen ***B***	Contains neither **anti-A** nor **anti-B** antibodies	 Contains both **anti-A** and **anti-B** antibodies

Blood Type	Frequency in Australia		Antigen	Antibody	Can donate blood to	Can receive blood from
	Rh^+	Rh^-				
A	31%	7%	A	anti-B	A, AB	A, O
B	8%	2%				
AB	2%	1%				
O	40%	9%				

Rh Blood Groups

Like the ABO grouping, the Rh system is based on antigens on the surfaces of red blood cells (RBCs). People whose RBCs have Rh antigens are Rh^+. Those who lack the Rh antigen are Rh^-. About 81% of people in Australia are Rh^+. Normally, human plasma does not contain antibodies against the Rh antigen but these may be produced during pregnancy if an Rh^- mother carries an Rh^+ fetus.

1. Complete the table above to show the antibodies and antigens in each blood group, and donor/recipient blood types:
2. Explain why blood from an incompatible donor causes a transfusion reaction in the recipient: _____
3. Why is blood type O^- sometimes called the universal donor? _____
4. Why is blood type AB^+ sometimes called the universal recipient? _____
5. Why was the discovery of the ABO system such a significant medical breakthrough? _____

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94 The Body's Defences

Key idea: The human body has a tiered system of defences against disease-causing organisms.

The body has several lines of defence against disease causing organisms (**pathogens**). The first line of defence consists of external barrier to stop pathogens entering the body. If this fails, a second line of defence targets any foreign bodies (including pathogens) that enter. Lastly, the immune system provides specific or targeted defence against the pathogen. The ability to ward off disease through the various defence mechanisms is called **resistance**. **Non-specific** (or innate) **resistance** protects against a broad range of pathogens and is provided by the first and second lines of defence. **Specific resistance** (the immune response) is the third tier of defence and is specific to a particular pathogen. Part of the immune response involves the production of **antibodies** (proteins that identify and neutralise foreign material). Antibodies recognise and respond to **antigens**, foreign or harmful substances that cause an immune response.

1st Line of Defence

The skin provides a physical barrier to the entry of pathogens. Healthy skin is rarely penetrated by microorganisms. Its low pH is unfavourable to the growth of many bacteria and its chemical secretions (e.g. sebum, antimicrobial peptides) inhibit growth of bacteria and fungi. Tears, mucus, and saliva also help to wash bacteria away.

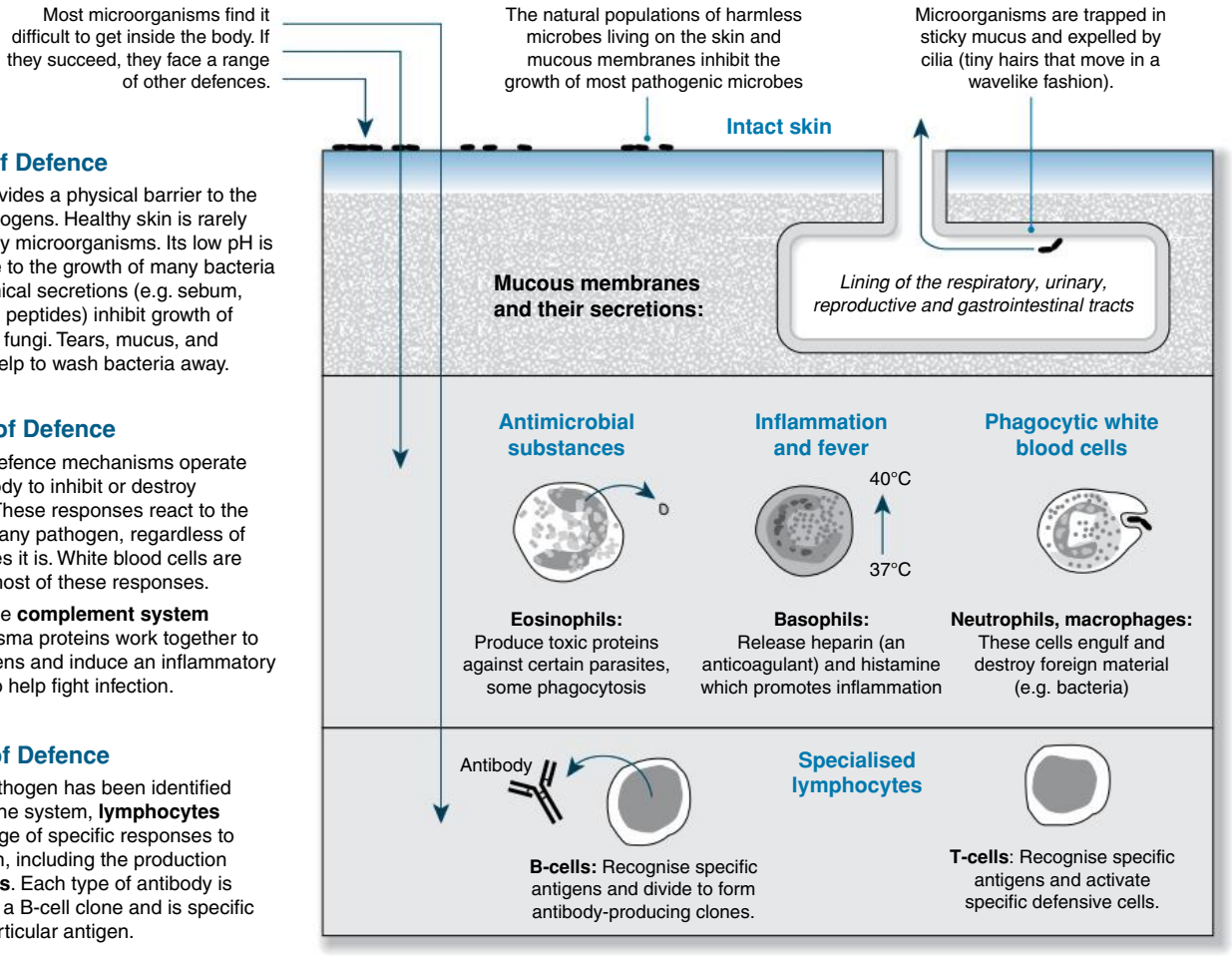
2nd Line of Defence

A range of defence mechanisms operate inside the body to inhibit or destroy pathogens. These responses react to the presence of any pathogen, regardless of which species it is. White blood cells are involved in most of these responses.

It includes the **complement system** whereby plasma proteins work together to bind pathogens and induce an inflammatory responses to help fight infection.

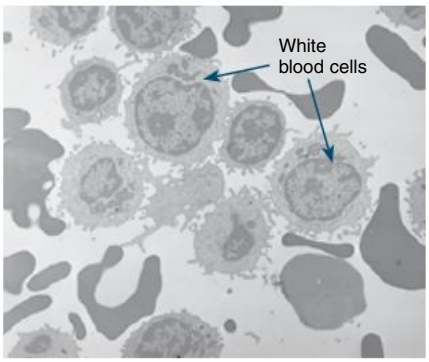
3rd Line of Defence

Once the pathogen has been identified by the immune system, **lymphocytes** launch a range of specific responses to the pathogen, including the production of **antibodies**. Each type of antibody is produced by a B-cell clone and is specific against a particular antigen.



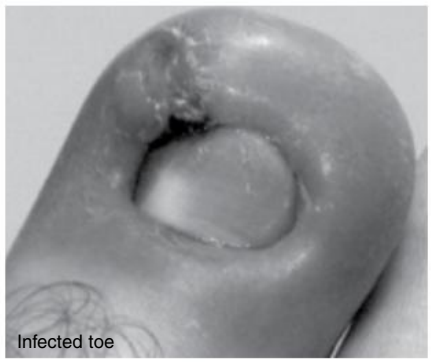


Tears contain antimicrobial substances as well as washing contaminants from the eyes.



EII

A range of white blood cells (arrowed above) form the second line of defence.



Wikimedia Commons

Inflammation is a localised response to infection characterised by swelling, pain, and redness.

1. Distinguish between specific and non-specific resistance: _____

_____

_____

_____

_____

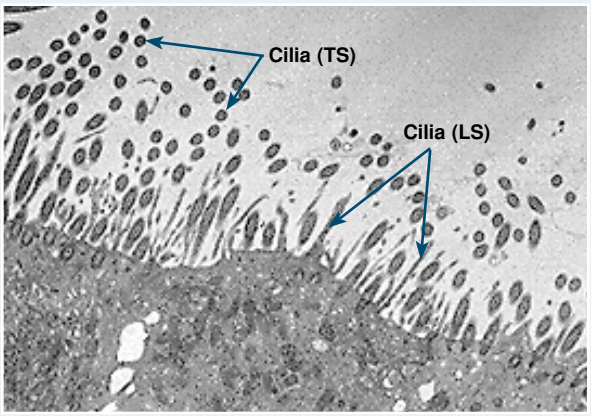
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The Importance of the First Line of Defence

The skin is the largest organ of the body. It forms an important physical barrier against the entry of pathogens into the body. A natural population of harmless microbes live on the skin, but most other microbes find the skin inhospitable. The continual shedding of old skin cells (arrow, right) physically removes bacteria from the surface of the skin. Sebaceous glands in the skin (top right) produce sebum, which has antimicrobial properties, and the slightly acidic secretions of sweat inhibit microbial growth.



Cilia line the epithelium of the **nasal passage** (below right). Their wave-like movement sweeps foreign material out and keeps the passage free of microorganisms, preventing them from colonising the body.



Antimicrobial chemicals are present in many bodily secretions. Tears, saliva, nasal secretions, and human breast milk all contain **lysozymes** and **phospholipases**. Lysozymes kill bacterial cells by catalysing the hydrolysis of cell wall linkages, whereas phospholipases hydrolyse the phospholipids in bacterial cell membranes, causing bacterial death. Low pH gastric secretions also inhibit microbial growth, and reduce the number of pathogens establishing colonies in the gastrointestinal tract.

2. How does the skin act as a barrier to prevent pathogens entering the body? _____

3. Describe the role of each of the following in non-specific defence:

   (a) Phospholipases: _____

   (b) Cilia: _____

   (c) Sebum: _____

4. Describe the functional role of each of the following defence mechanisms:

   (a) Phagocytosis by white blood cells: _____

   (b) Antimicrobial substances: _____

   (c) Antibody production: _____

5. Explain the value of a three tiered system of defence against microbial invasion: _____

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95 The Action of Phagocytes

Key Idea: Phagocytes are types of white blood cells that ingest microbes and digest them by phagocytosis.

All types of **phagocytes** are white blood cells. They ingest microbes and digest them by **phagocytosis.** During many kinds of infections, especially bacterial infections, the total number of white blood cells increases by two to four times the normal number. The ratio of various white blood cell types changes during the course of an infection.

How a Phagocyte Destroys Microbes

1. **Detection**
Phagocyte detects microbes by the chemicals they give off (chemotaxis) and sticks the microbes to its surface.

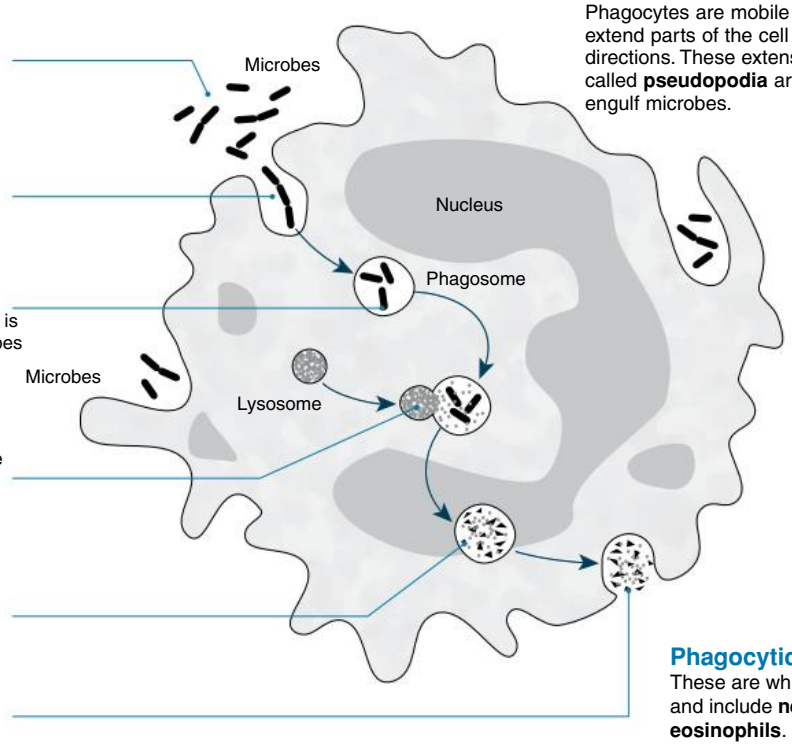
2. **Ingestion**
The microbe is engulfed by the phagocyte wrapping pseudopodia around it to form a vesicle.

3. **Phagosome forms**
A phagosome (phagocytic vesicle) is formed, which encloses the microbes in a membrane.

4. **Fusion with lysosome**
Phagosome fuses with a lysosome (which contains powerful enzymes that can digest the microbe).

5. **Digestion**
The microbes are broken down by enzymes into their chemical constituents.

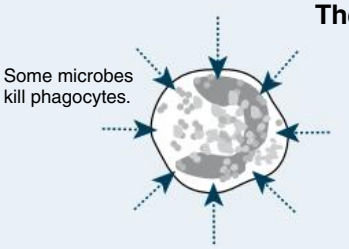
6. **Discharge**
Indigestible material is discharged from the phagocyte cell.



Phagocytes are mobile cells that can extend parts of the cell in different directions. These extensions are called **pseudopodia** are used to engulf microbes.

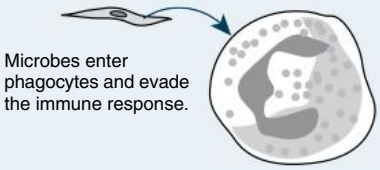
Phagocytic cell
These are white blood cells and include **neutrophils** and **eosinophils**.

The Interaction of Microbes and Phagocytes



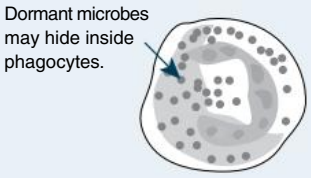
Some microbes kill phagocytes

Some microbes produce toxins that can actually kill phagocytes, e.g. toxin-producing staphylococci and the dental plaque-forming bacteria *Actinobacillus*.



Microbes evade immune system

Some microbes can evade the immune system by entering phagocytes. The microbes prevent fusion of the lysosome with the phagosome and multiply inside the phagocyte, almost filling it. Examples include *Chlamydia*, *Mycobacterium tuberculosis*, *Shigella*, and malarial parasites.



Dormant microbes hide inside

Some microbes can remain dormant inside the phagocyte for months or years at a time. Examples include the microbes that cause brucellosis and tularemia.

1. Identify the white blood cells capable of phagocytosis: _____

_____

2. How can a blood sample be used to diagnose a microbial infection (without looking for the microbes themselves)?

_____

_____

3. Explain how some microbes can overcome phagocytic cells and use them to their advantage: _____

_____

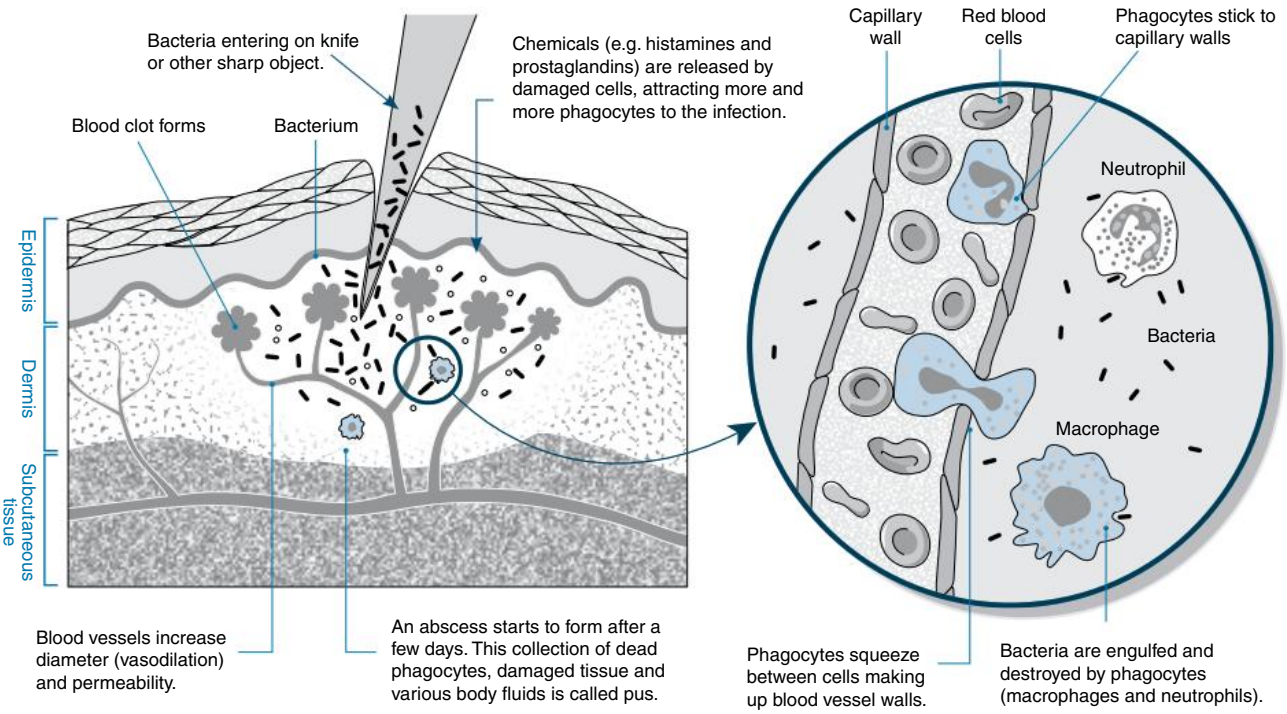
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96 Inflammation

Key Idea: Inflammation is a type of non-specific resistance in response to harmful stimuli, such as pathogens.

Damage to the body's tissues (e.g. by sharp objects, heat, or microbial infection) triggers a defensive response called **inflammation**. It is usually characterised by four symptoms: pain, redness, heat, and swelling. The inflammatory response is beneficial and has the following functions: (1) to destroy the cause of the infection and remove it and its products from the body; (2) if this fails, to limit the effects on the body by confining the infection to a small area; (3) replacing or repairing tissue damaged by the infection. Inflammation can be divided into three distinct phases, shown below.



Stages in inflammation

Increased diameter and permeability of blood vessels
Blood vessels increase their diameter and permeability in the area of damage. This increases blood flow to the area and allows defensive substances to leak into tissue spaces.

→

Phagocyte migration and phagocytosis
Within one hour of injury, phagocytes appear on the scene. They squeeze between cells of blood vessel walls to reach the damaged area where they destroy invading microbes.

→

Tissue repair
Functioning cells or supporting connective cells create new tissue to replace dead or damaged cells. Some tissue regenerates easily (skin) while others do not at all (cardiac muscle).

1. Outline the three stages of inflammation and identify the beneficial role of each stage:

   (a) _____

   _____

   (b) _____

   _____

   (c) _____

   _____

2. Identify two features of phagocytes important in the response to microbial invasion: _____

   _____

3. What is the role of histamines and prostaglandins in inflammation? _____

4. Why does pus form at the site of infection? _____

   _____

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LINK 107 LINK 94 WEB 96 KNOW

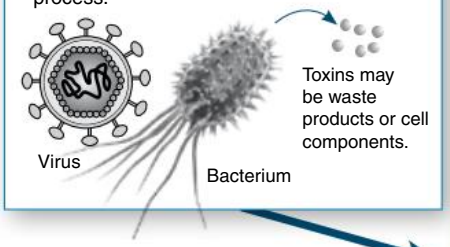
97 Fever

Key Idea: Fever is the rise in body temperature caused by the resetting the of body's temperature set-point in response to an infection.

Fever describes an increase in internal body temperature above the normal 36.2°C-37.2°C. It arises because of an increase in the body's thermoregulatory set-point so that the previous "normal body temperature" is considered low (**hypothermic**). Fever is not a disease, but it is a symptom of infection. Fever is helpful in some ways. The release of the protein **interleukin-1** helps to reset the thermostat of the body to a higher level, and increases production of **T cells** (lymphocytes). Fever also intensifies the effect of **interferon** (an antiviral protein) and may slow bacterial growth and viral replication. It also speeds up **metabolism**, promoting more rapid tissue repair. Fever also increases heart rate so white blood cells can be delivered to sites of infection more rapidly.

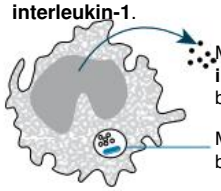
1 Pathogen or toxin

The most frequent cause of fever is infection from bacteria (and their toxins) and viruses. A macrophage ingesting one of these will start the fever-causing process.



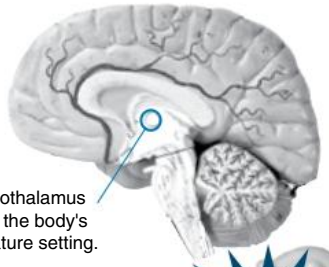
2 Macrophages respond

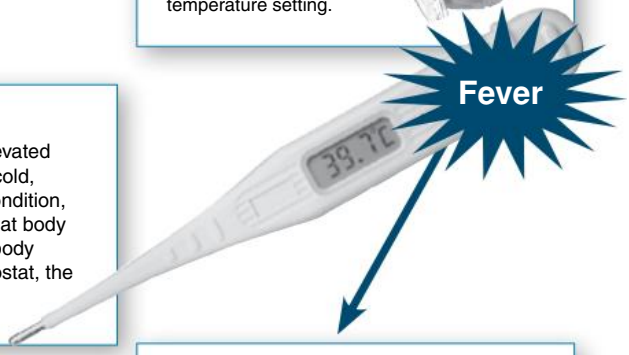
Macrophage ingests bacterium, destroying it and releasing endotoxins. The endotoxins induce the macrophage to produce a small protein called **interleukin-1**.



3 Thermostat is reset

Interleukin-1 circulates to the brain and induces the hypothalamus to produce more prostaglandins, which resets the 'thermostat' to a **higher temperature**, producing fever.





4 Fever onset

The body responds to a higher temperature set point by constricting blood vessels, increasing metabolic rate, and **shivering**. These responses raise body temperature beyond the normal range of 36.2 – 37.2 °C

5 Chill phase

Although body temperature is elevated above normal, the skin remains cold, and **shivering** increases. This condition, called a **chill**, is a definite sign that body temperature is rising. When the body reaches the setting of the thermostat, the chill disappears.



We are all familiar with the feeling of chill associated with a fever and the sweating that precedes the fever's resolution. Fevers of less than 40°C do not need treatment but high, prolonged fevers require prompt attention, because death usually results if the body temperature rises above 45.5°C.

6 Crisis phase

Body temperature is maintained at the higher setting until the interleukin-1 has been eliminated. As the infection subsides, the **thermostat is reset to 37°C**. Heat losing mechanisms, such as sweating and vasodilation cause the person to feel warm. This **crisis phase** of the fever indicates that body temperature is falling.

1. Discuss the beneficial effects of fever on the body's ability to fight infections: _____

2. Summarise the key steps of how the body's thermostat is set at a higher level by infection: _____

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98 The Lymphatic System

Key Idea: The lymphatic system collects the fluid leaked from the blood vessels and returns it to the heart.

The body's tissues are bathed in a plasma-like fluid called tissue fluid, which supplies them with nutrients and oxygen, and removes wastes. Tissue fluid is similar to plasma, although it lacks large proteins, and it is formed when fluid leaks out from capillaries. Most of the tissue fluid is returned to the blood vessels, but about 3 L per day is collected by the lymph vessels of the lymphatic system and returned to the heart. The **lymphatic system**, which is made up of the **lymph**, lymph vessels and lymph tissues (below) has an important role in role in internal defence. Lymph nodes are the main sites where pathogens are destroyed and become swollen when fighting an infection. The thymus, spleen, and bone marrow contribute leucocytes to the lymphatic and circulatory systems.

Tonsils: Tonsils (and adenoids) comprise a collection of large lymphatic nodules at the back of the throat. They produce lymphocytes and antibodies and are well-placed to protect against invasion of pathogens.

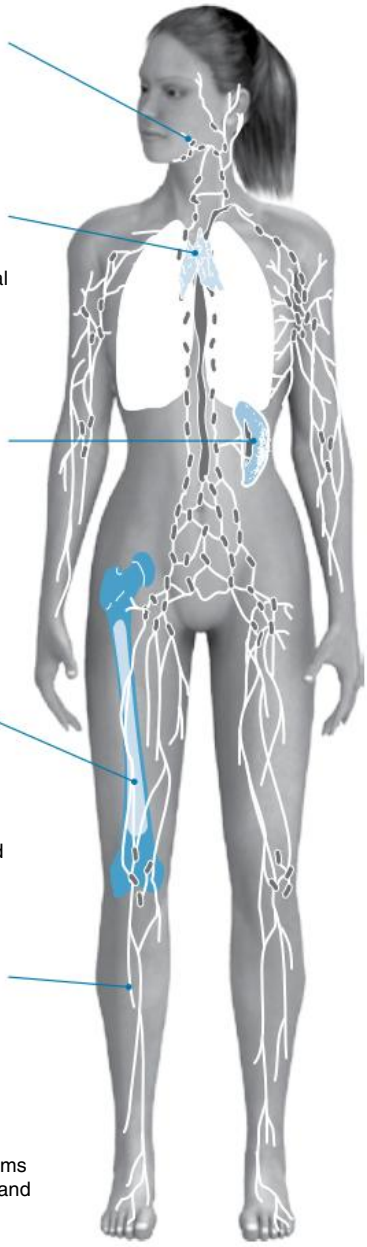
Thymus gland: The thymus is a two-lobed organ located close to the heart. It is large in infants and shrinks after puberty to a fraction of its original size. It helps produce the **T-cells** that destroy invading microbes.

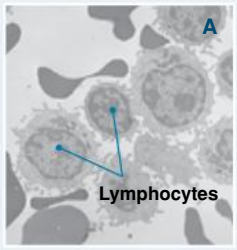
Spleen: The oval spleen is the largest mass of lymphatic tissue in the body. It stores and releases blood in case of demand (e.g. in cases of bleeding), produces mature **B-cells**, and destroys bacteria by phagocytosis.

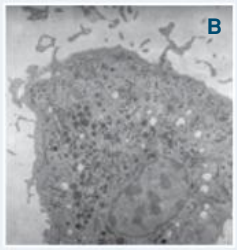
Bone marrow: All the different kinds of blood cells are produced by cellular differentiation from stem cells in the red bone marrow.

Lymphatic vessels: When tissue fluid is picked up by lymph capillaries, it is called **lymph**. The lymph is passed along lymphatic vessels to a series of lymph nodes. These vessels contain one-way valves that move the lymph back to the heart where it re-enters the general circulation.

Disorders of the lymphatic system: If the lymphatic system is not working properly, fluids can build up causing swelling (lymphoedema). Other problems can result from infections, blockages, and cancer (e.g. Hodgkin's disease).



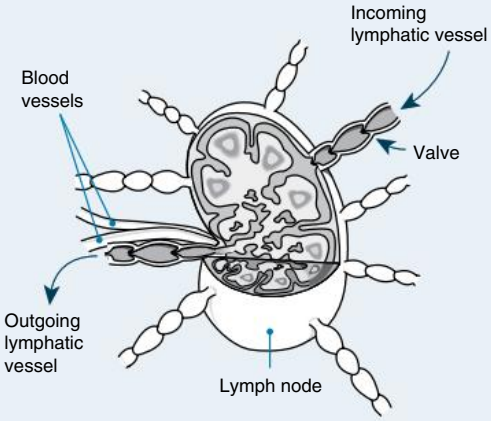




Photos: EII

Many types of leucocytes are involved in internal defence. The photos above illustrate examples of leucocytes. Photo **A** shows a cluster of **lymphocytes**. Photo **B** shows a single **macrophage**: large, phagocytic cells that develop from monocytes and move from the blood to reside in many organs and tissues, including the spleen and lymph nodes.

Lymph node



Lymph nodes are oval or bean-shaped structures, scattered throughout the body, usually in groups, along the length of lymphatic vessels. As lymph passes through the nodes, it filters foreign particles (including pathogens) by trapping them in fibres. Lymph nodes are also a "store" of **lymphocytes**, which may circulate to other parts of the body. Once trapped, macrophages destroy the foreign substances by phagocytosis. T cells may destroy them by releasing various products, and/or B-cells may release antibodies that destroy them.

1. Briefly describe the composition of **lymph**: _____

_____

2. Discuss the various roles of lymph: _____

_____

_____

3. State one role of each of the following in the lymphatic system:

   (a) Lymph nodes: _____

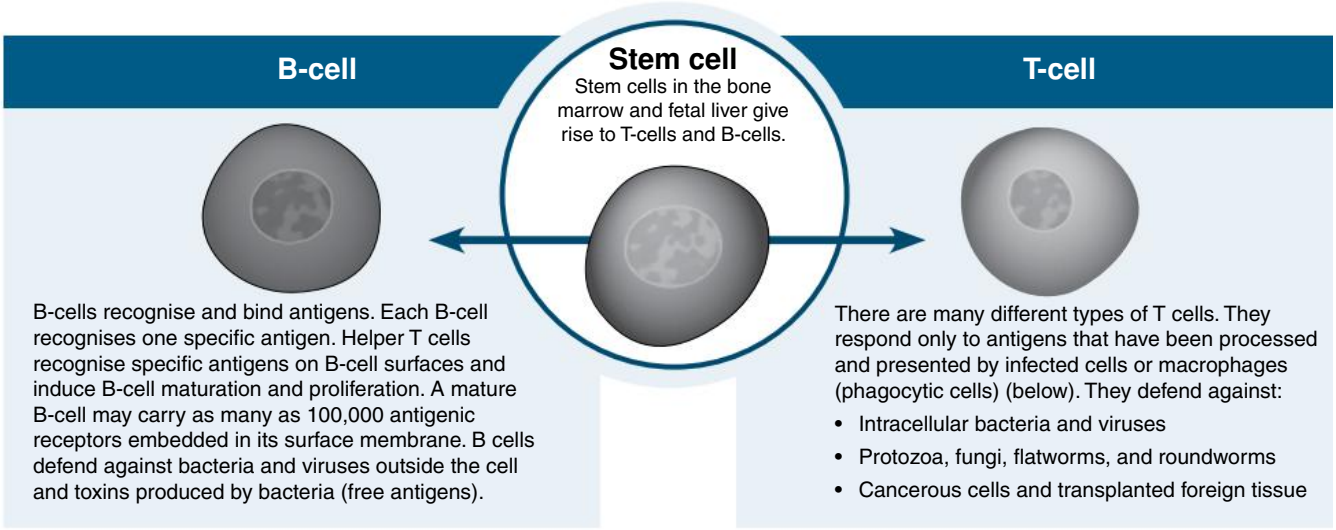
   (b) Bone marrow: _____

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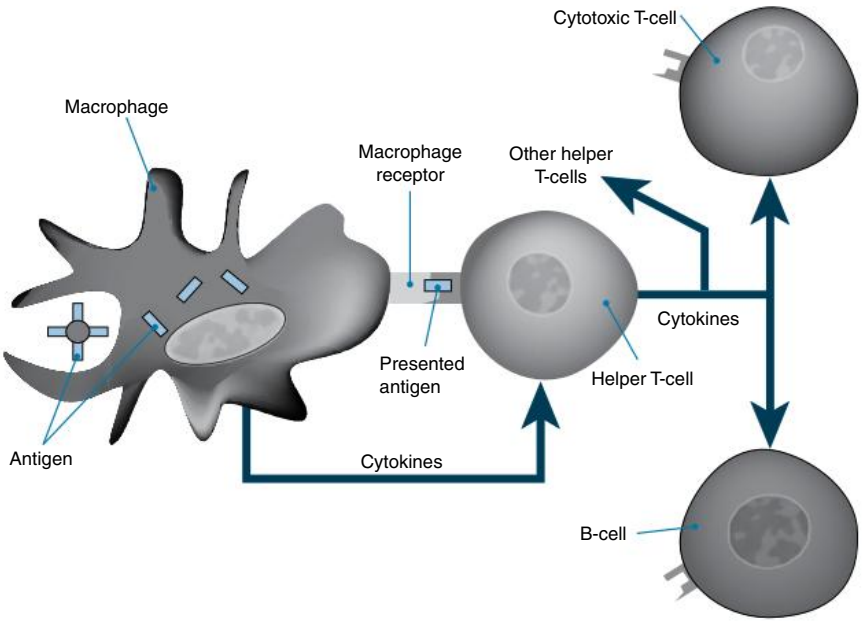
99 The Immune System

Key Idea: The defence provided by the immune system is based on its ability to respond specifically against foreign substances and hold a memory of this response.

There are two main components of the immune system: the humoral and the cell-mediated responses. They work separately and together to provide protection against disease. The **humoral immune response** is associated with the serum (the non-cellular part of the blood) and involves the action of antibodies secreted by B-cell lymphocytes. Antibodies are found in extracellular fluids including lymph, plasma, and mucus secretions. They protect the body against viruses, and bacteria and their toxins. The **cell-mediated immune response** is associated with the production of specialised lymphocytes called **T-cells**.



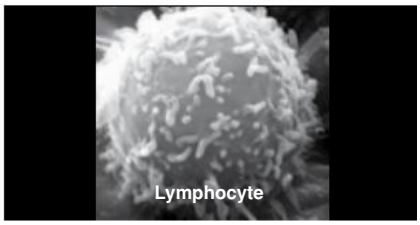
B Cell and T Cell Activation



Helper T-cells are activated by direct cell-to-cell signalling and by signalling to nearby cells using **cytokines** from macrophages.

Macrophages ingest antigens, process them, and present them on the cell surface where they are recognised by helper T-cells. The helper T-cell binds to the antigen and to the macrophage receptor, which leads to activation of the helper T-cell.

The macrophage also produces and releases cytokines, which enhance T-cell activation. The activated T-cell then releases more cytokines which causes the proliferation of other helper T-cells (positive feedback) and helps to activate cytotoxic T-cells and antibody-producing B-cells.



1. Describe the general action of the two major divisions in the immune system:

   (a) Humoral immune system: _____

   _____

   (b) Cell-mediated immune system: _____

   _____

2. Explain how an antigen causes the activation and proliferation of T-cells and B-cells: _____

   _____

   _____

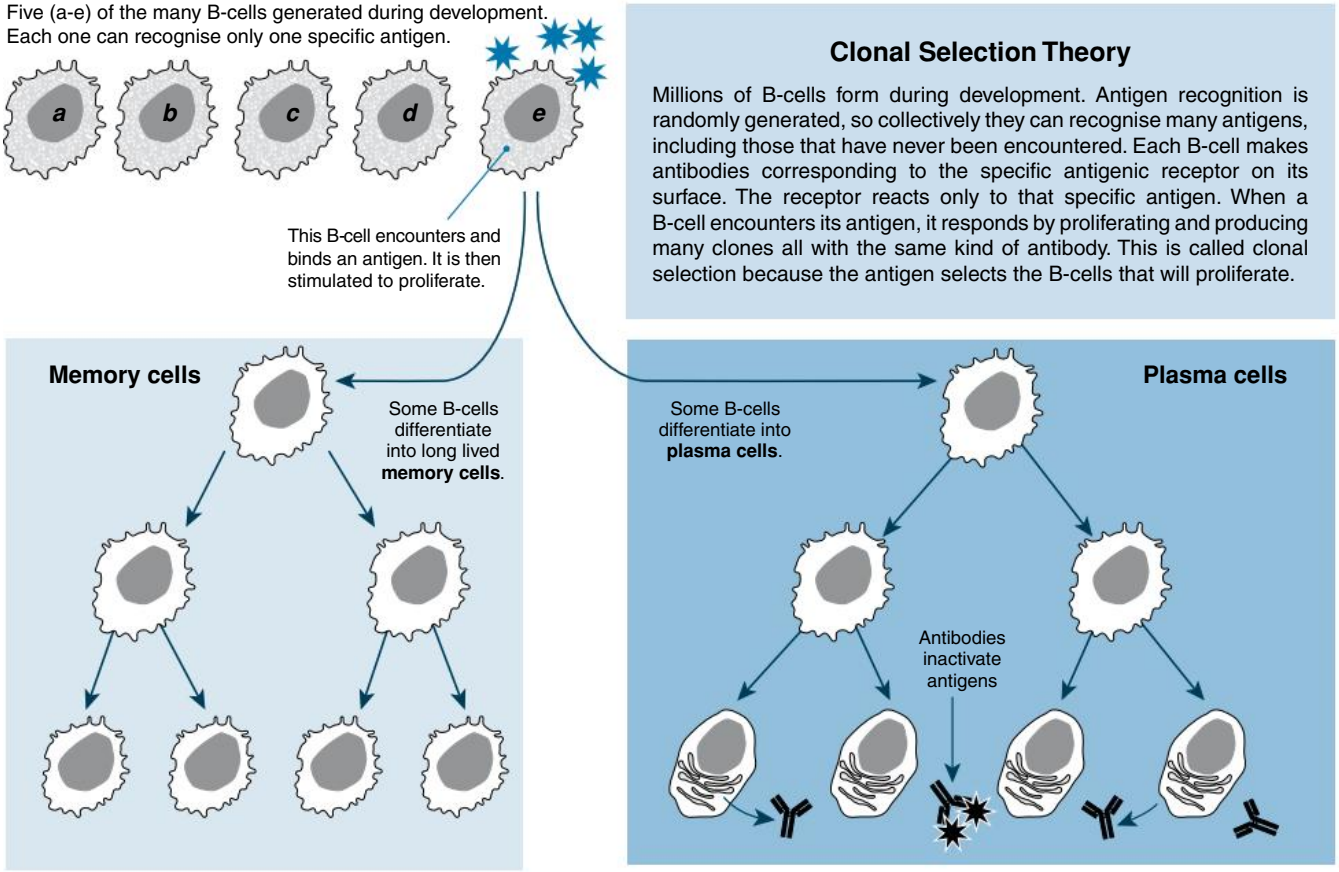
   _____

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100 Clonal Selection

Key Idea: Clonal selection theory explains how lymphocytes can respond to a large and unpredictable range of antigens.

In 1955, the Australian, **Sir Frank Macfarlane Burnet** proposed the **clonal selection theory** to explain the response of the immune system to the wide range of potential antigens in the environment. The diagram below describes **clonal selection** in B-cells after antigen exposure. In the same way, a T-cell stimulated by a specific antigen will multiply and develop into different types of T-cells. Clonal selection and differentiation of lymphocytes provide the basis for **immunological memory**.



Some B-cells differentiate into long lived **memory cells**. These are retained in the lymph nodes to provide future immunity (**immunological memory**). In the event of a second infection, B-memory cells react more quickly and vigorously than the initial B-cell reaction to the first infection.

Plasma cells secrete antibodies specific to the antigen that stimulated their development. Each plasma cell lives for only a few days, but can produce about 2000 antibody molecules per second. Note that during development, any B-cells that react to the body's own antigens are selectively destroyed in a process that leads to **self tolerance** (acceptance of the body's own tissues).

1. Describe how clonal selection results in the proliferation of one particular B-cell: _____

2. Describe the function of each of the following cells in the immune system response:

   (a) Memory cells: _____

   (b) Plasma cells: _____

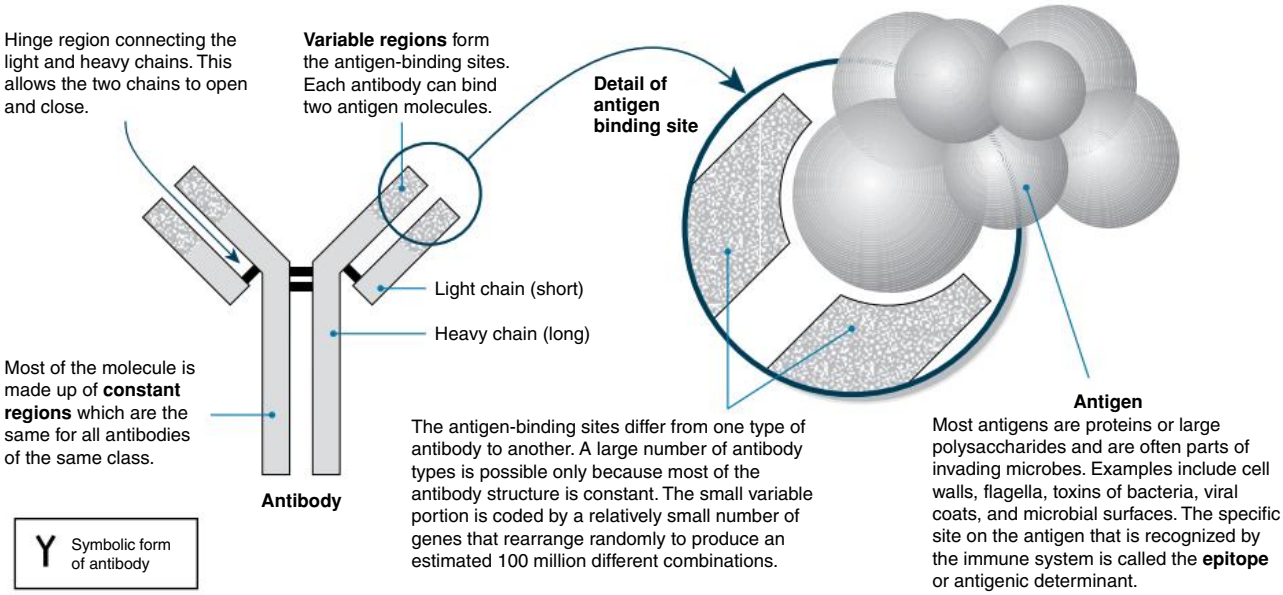
3. Explain the basis of **immunological memory**: _____

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101 Antibodies

Key Idea: Antibodies are large, Y-shaped proteins, made by plasma cells, which destroy specific antigens.

Antibodies and antigens play key roles in the response of the immune system. **Antigens** are foreign molecules that are able to bind to antibodies (or T cell receptors) and provoke a specific immune response. Antigens include potentially damaging microbes and their toxins, as well as substances such as pollen grains, cell surface proteins. **Antibodies** (or immunoglobulins) are proteins made in response to antigens. They are secreted into the plasma where they can recognise, bind to, and help to destroy antigens. There are five classes of **immunoglobulins**. Each plays a different role in the immune response (e.g. enhancing phagocytosis or neutralising toxins). Each type of antibody is specific to only one particular antigen. The ability of the immune system to recognise and ignore the antigenic properties of its own tissues occurs early in development and is called **self-tolerance**. Exceptions occur when the immune system malfunctions and the body attacks its own tissues, causing an **autoimmune disorder**.



1. Distinguish between an **antibody** and an **antigen**: _____

2. Describe the structure of an antibody, identifying the specific features of its structure that contribute to its function:

3. It is necessary for the immune system to clearly distinguish the body's own cells and proteins from foreign ones.

   (a) Why is this the case? _____

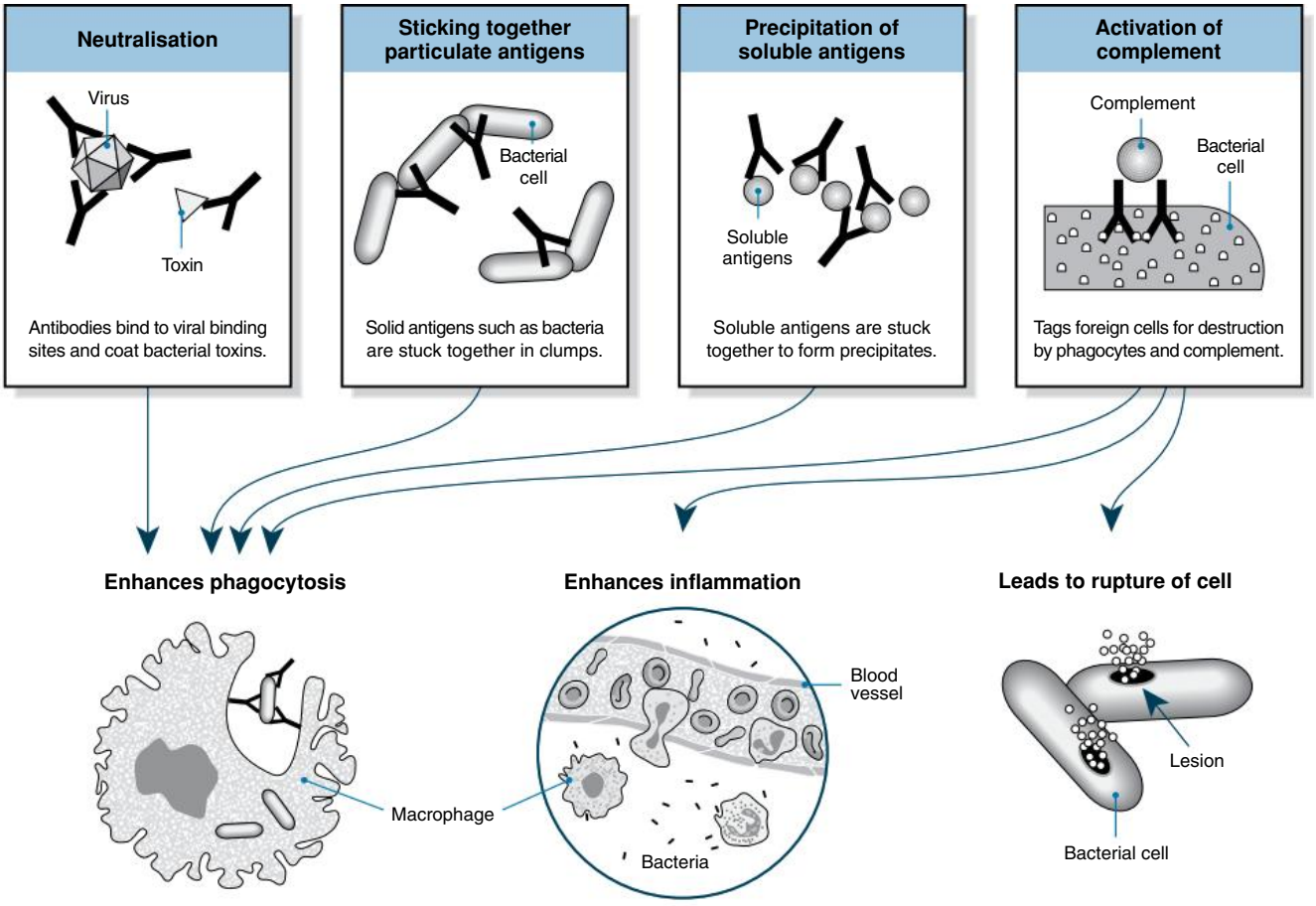
   (b) How does **self tolerance** develop? _____

   (c) What type of disorder results when this recognition system fails? _____

   (d) Describe two examples of disorders that are caused in this way, identifying what happens in each case:

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How Antibodies Inactivate Antigens



4. Discuss the various ways in which antibodies inactivate antigens: _____

5. Explain how antibody activity enhances or leads to:

   (a) Phagocytosis: _____

   (b) Inflammation: _____

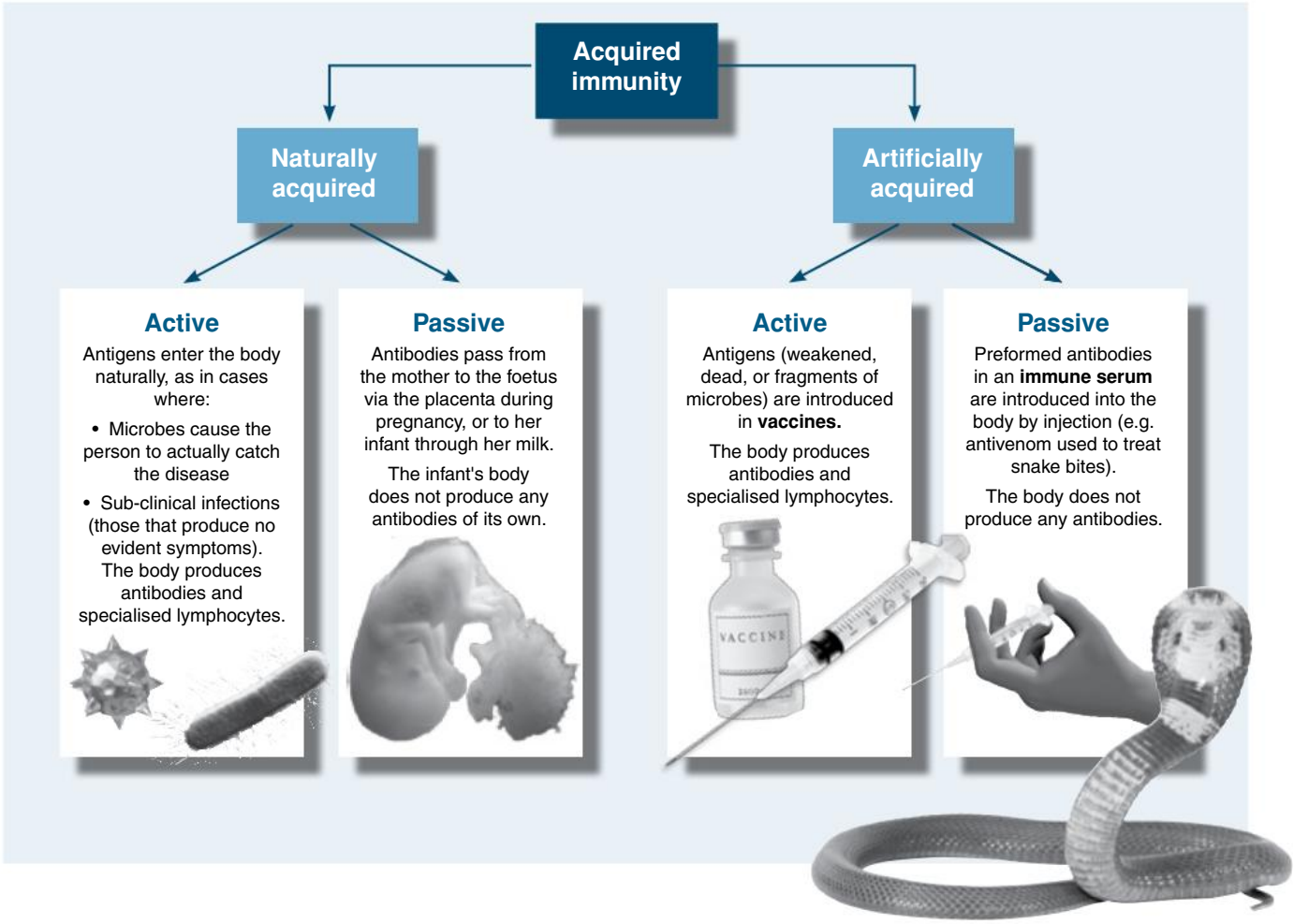
   (c) Bacterial cell lysis: _____

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102 Acquired Immunity

Key Idea: Acquired immunity is a resistance to specific pathogens acquired over the life-time of an organism.

We are born with natural or **innate resistance** which provides non-specific immunity to certain illnesses. In contrast, **acquired immunity** is protection developed over time to specific antigens. **Active immunity** develops after the immune system responds to being exposed to microbes or foreign substances. **Passive immunity** is acquired when antibodies are transferred from one person to another. Immunity may also be naturally acquired, through natural exposure to microbes, or artificially acquired as a result of medical treatment (below).



1. (a) What is meant by **passive immunity**? _____

_____

(b) Distinguish between naturally and artificially acquired passive immunity and give an example of each:

_____

_____

2. (a) Why does a newborn baby need to have received a supply of maternal antibodies prior to birth? _____

_____

_____

(b) Why is this supply supplemented by antibodies in breast milk? _____

_____

(c) Would you recommend breast feeding to a new mother? Explain your answer: _____

_____

_____

_____

KNOW LINK 101 LINK 103

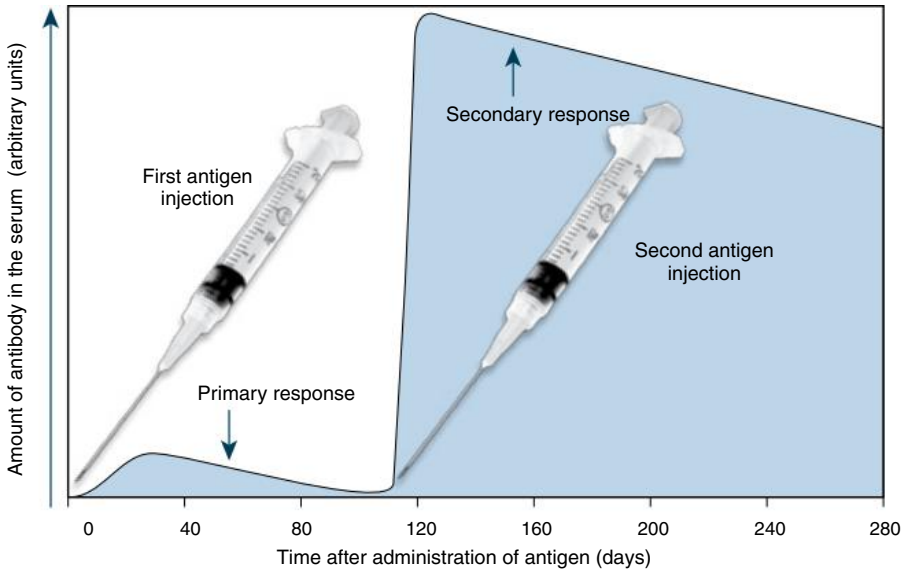
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Primary and Secondary Responses to Antigens

When the B cells encounter antigens and produce antibodies, the body develops **active immunity** against that antigen.

The initial response to antigenic stimulation, caused by the sudden increase in B cell clones, is called the **primary response**. Antibody levels as a result of the primary response peak a few weeks after the response begins and then decline. However, because the immune system develops an immunological memory of that antigen, it responds much more quickly and strongly when presented with the same antigen subsequently (the **secondary response**).

This forms the basis of immunisation programmes where one or more booster shots are provided following the initial vaccination.





Vaccines against common diseases are given at various stages during childhood according to an immunisation schedule. Vaccination has been behind the decline of some once-common childhood diseases, such as mumps.



Many childhood diseases for which vaccination programmes exist are kept at a low level because of **herd immunity**. If most of the population is immune, those that are not immunised may be protected because the disease is uncommon.



CDC

Most vaccinations are given in childhood, but adults may be vaccinated against a disease (e.g. TB, influenza) if they are in a high risk group (e.g. the elderly) or if they are travelling to a region in the world where a disease is prevalent.

3. (a) What is **active immunity**? _____

(b) Distinguish between naturally and artificially acquired active immunity and give an example of each: _____

4. (a) Describe two differences between the primary and secondary responses to presentation of an antigen: _____

(b) Why is the secondary response so different from the primary response? _____

5. (a) Explain the principle of **herd immunity**: _____

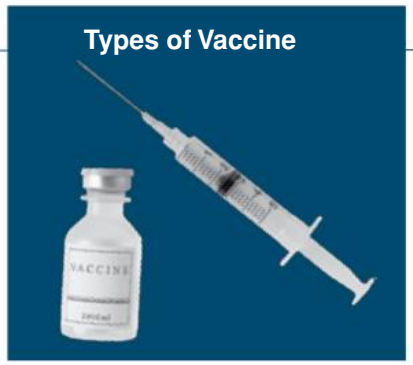
(b) Why are health authorities concerned when the vaccination rates for an infectious disease fall?

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103 Vaccines and Vaccination

Key Idea: A vaccine is a suspension of microorganisms (or pieces of them) that is deliberately introduced into the body to protect against disease. It induces immunity by stimulating the production of antibodies.

A **vaccine** is a preparation of a harmless foreign antigen that is deliberately introduced into the body to produce an immune response. The antigen in the vaccine triggers the immune system to produce antibodies against the antigen, but it does not cause the disease. The immune system remembers its response and will produce the same antibodies if it encounters the antigen again. There are two basic types of vaccine, subunit vaccines and whole-agent vaccines. **Whole-agent vaccines** contain complete non-virulent microbes, either inactivated (killed), or alive but attenuated (weakened). Attenuated vaccines are the most effective of these and often provide life-long immunity. **Subunit vaccines** contain parts of the pathogen that induce an immune response. They are safer than attenuated vaccines because they cannot reproduce in the recipient, and they produce fewer adverse effects because they contain little or no extra material.



Whole-Agent Vaccine

Contains whole, **nonvirulent** microorganisms

Subunit Vaccine

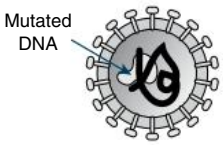
Contains some part or product of microorganisms that can produce an immune response

Inactivated (killed)



Viruses for vaccines may be inactivated with formalin or other chemicals. They present no risk of infection, e.g. most influenza vaccines, Salk polio vaccine.

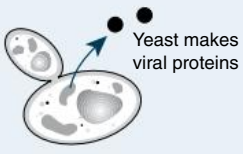
Attenuated (weakened)



Attenuated viruses are usually strains in which mutations have accumulated during culture. These live viruses can back-mutate to a virulent form, e.g. MMR vaccine.

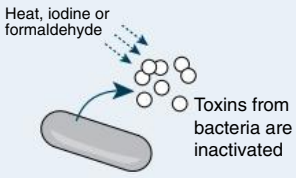
Many subunit vaccines are made with the addition of adjuvants: additional agents that increase the effectiveness of the vaccine. Adjuvants are not part of the pathogen but include irritants that increase the level of immune response.

Recombinant vaccines



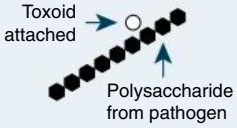
Non-pathogenic microbes are genetically engineered to make an antigenic fraction, e.g. the hepatitis B vaccine.

Toxoids



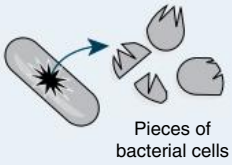
Bacterial toxins inactivated by heat or chemicals. The toxoid stimulates the production of antibodies, e.g. diphtheria vaccine, tetanus vaccine.

Conjugated vaccines



Polysaccharides from the pathogen are combined with proteins from other pathogens, e.g. *vaccine against Haemophilus* influenzae b.

Acellular vaccines

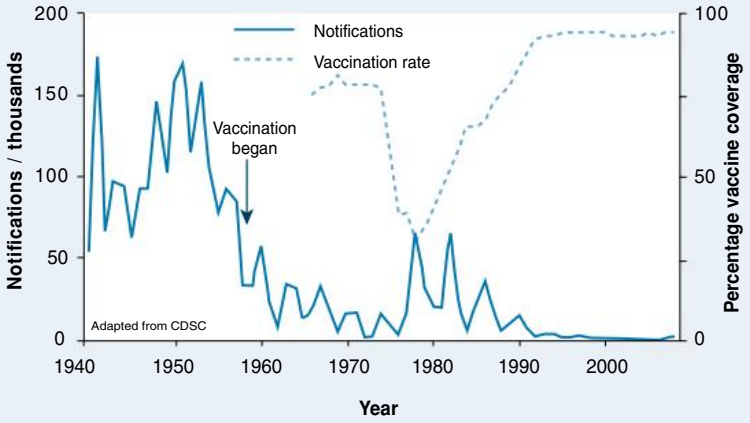


Fragments of a whole-agent vaccine containing the desired antigens. Unable to cause infection, e.g. typhoid vaccine.

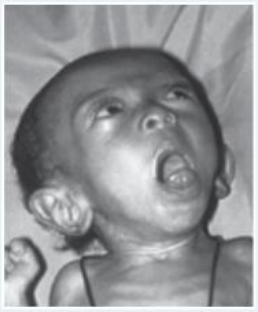
1. **Attenuated viruses** provide long term immunity to their recipients and generally do not require booster shots. Why do you think attenuated viruses provide such effective long-term immunity when inactivated viruses do not?

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Whooping cough notifications and vaccine coverage (England and Wales) 1940-2008



Whooping cough is caused by the bacterium *Bordetella pertussis*, and may last for two to three months. It is characterised by a whooping cough and painful coughing spasms, which may be followed by periods of vomiting. Infants under six months of age are most at risk of developing complications or dying because they are too young to be fully protected by the vaccine. Inclusion of the whooping cough vaccine into the UK immunisation schedule has greatly reduced the incidence rates of the disease (above).



CDC

Smallpox Eradication

Smallpox is a highly contagious disease caused by the *Variola* virus. It has two forms, the more severe of which kills about 30% of the people infected. Over several centuries, many smallpox epidemics swept across the globe, killing millions and maiming millions more. Survivors were often left blind and severely scarred. In the late 1700s, Edward Jenner discovered that inoculation with the cowpox virus could protect humans from smallpox.

Several countries began vaccination programmes, and during the 1960s the World Health Organisation led a global programme that resulted in the complete eradication of smallpox. The key to the strategy was **surveillance** of outbreaks and then **containment**. Eradication was helped by the fact that humans were the only reservoir for infection and there were no carriers. The last natural outbreak was recorded in Somalia in 1977.



CDC

The Bangladeshi girl (above) shows the typical smallpox lesion pattern on her face and torso. The smallpox vaccine (left) contains a live vaccinia virus. Vaccina virus is similar to the virus that causes smallpox. Stocks are kept in the Netherlands.

2. (a) How can vaccination help lead to the eradication of an infectious disease? _____

(b) Discuss factors in the success of the smallpox eradication programme: _____

3. (a) What factors would make eradication by vaccination difficult to achieve? _____

(b) Can you think of a disease that would be difficult to eradicate by vaccination? Explain your choice:

4. In 1975, the UK vaccination rate for whooping cough decreased to 30% because of concerns about the vaccine's safety. Use the data in the graph above to describe what effect this had on rates of whooping cough reported:

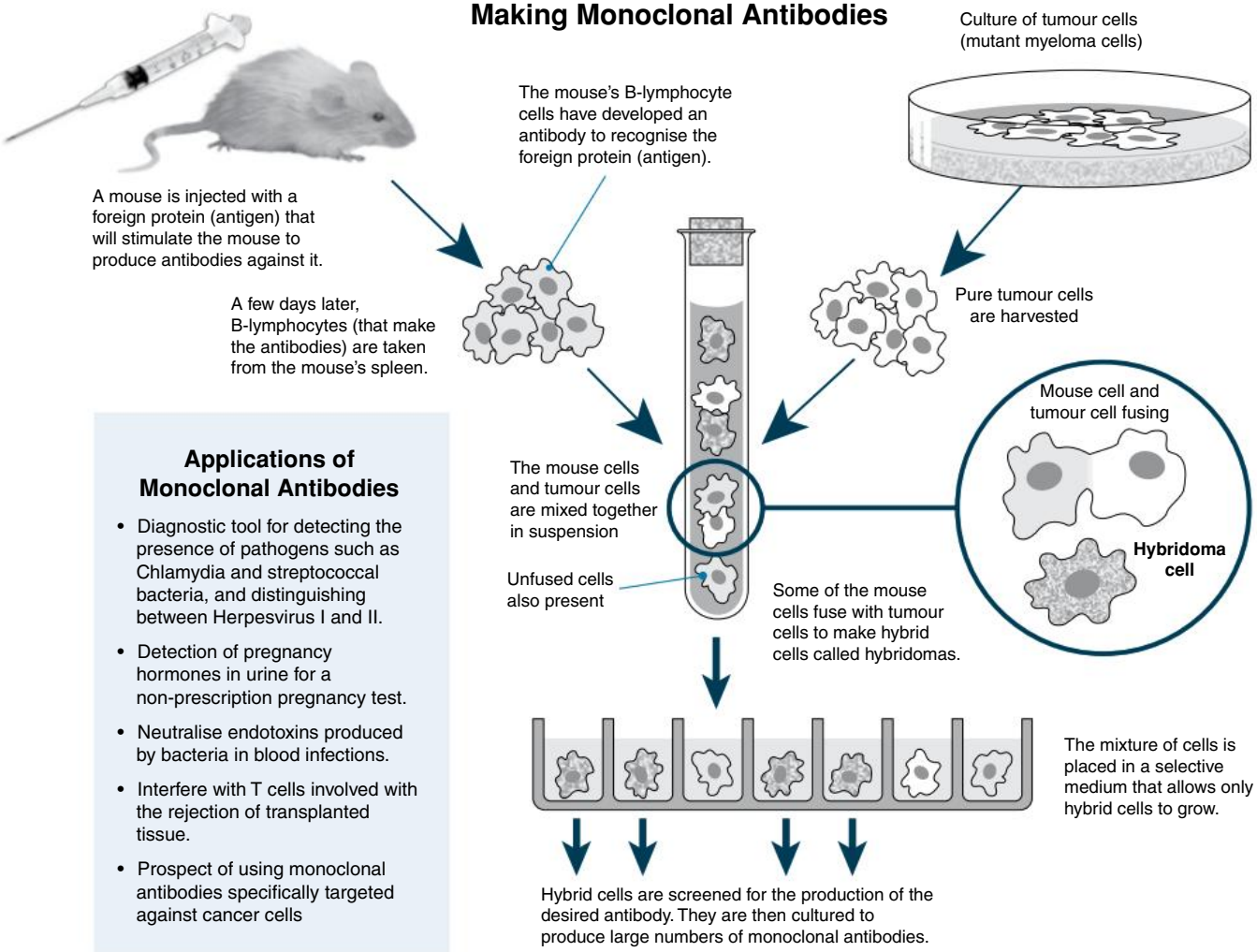
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104 Monoclonal Antibodies

Key Idea: Monoclonal antibodies are artificially produced antibodies that neutralise specific antigens. They have wide applications in diagnosing and treating disease, in detecting pregnancy, and in food safety tests.

A monoclonal antibody is an artificially produced antibody that binds to and neutralises one specific type of antigen. A monoclonal antibody binds an antigen in the same way that a normally produced antibody does. Monoclonal antibodies are produced by stimulating the production of antibody-producing B-cells in mice injected with the antigen. The isolated B-cells are made to fuse with immortal tumour cells, and can be cultured indefinitely in a suitable growing medium. Monoclonal antibodies are useful because they are identical (i.e. clones), they can be produced in large quantities, and they are highly specific. However, the immune systems of some people react against the foreign mouse proteins and results in an immune response. Antibodies produced in this way are used in medical diagnosis and treatment.

Making Monoclonal Antibodies



Applications of Monoclonal Antibodies

- Diagnostic tool for detecting the presence of pathogens such as Chlamydia and streptococcal bacteria, and distinguishing between Herpesvirus I and II.
- Detection of pregnancy hormones in urine for a non-prescription pregnancy test.
- Neutralise endotoxins produced by bacteria in blood infections.
- Interfere with T cells involved with the rejection of transplanted tissue.
- Prospect of using monoclonal antibodies specifically targeted against cancer cells

1. (a) Which mouse cells are used to produce monoclonal antibodies? _____

   (b) What problem is associated with the use of mice to produce monoclonal antibodies? _____

   _____

2. Which characteristic of tumour cells allows an ongoing culture of antibody-producing lymphocytes to be made?

   _____

   _____

3. Describe four applications of monoclonal antibodies:

   (a) _____

   (b) _____

   (c) _____

   (d) _____

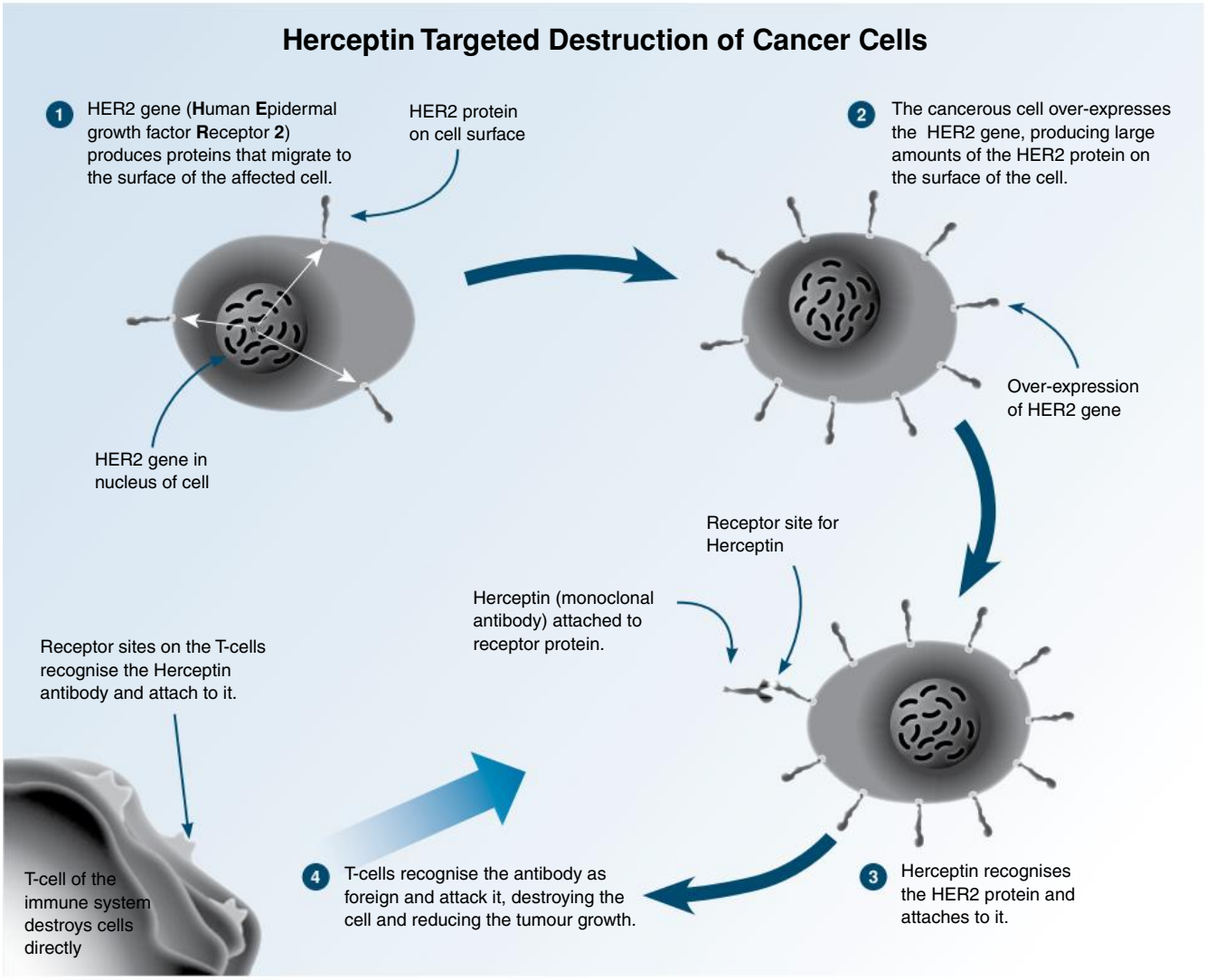
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105 Herceptin: A Modern Monoclonal

Key Idea: Herceptin is a monoclonal antibody that attaches to the HER2 receptor protein on cells to help T-cells target them for destruction.

Herceptin is the patented name of a monoclonal antibody for the treatment of breast cancer. It targets the HER2 receptor proteins on cancerous cells that signal to the cell when it should divide. The proteins are produced by the **proto-oncogene HER2**. Cancerous cells contain 20-30% more of the HER2 gene than normal cells and this causes **over-expression** of HER2, and large amounts of HER2 protein. The over-expression causes the cell to divide more often than normal, producing a tumour. Cancerous (**HER2⁺**) cells are not recognised by the immune system as abnormal. Herceptin recognises and binds to the HER2 protein on the surface of the cancerous cell so that the immune system can recognise it as foreign and destroy it.

Herceptin Targeted Destruction of Cancer Cells



1. (a) Why does the over-expression of the HER2 gene cause cancer? _____

_____

_____

(b) Why does the immune system require Herceptin to help target cancerous cells? _____

_____

2. Describe the two ways in which Herceptin works to fight cancer:

(a) _____

(b) _____

3. Why is the use of Herceptin and other monoclonal antibody drugs called **targeted drug therapy**? _____

_____

_____

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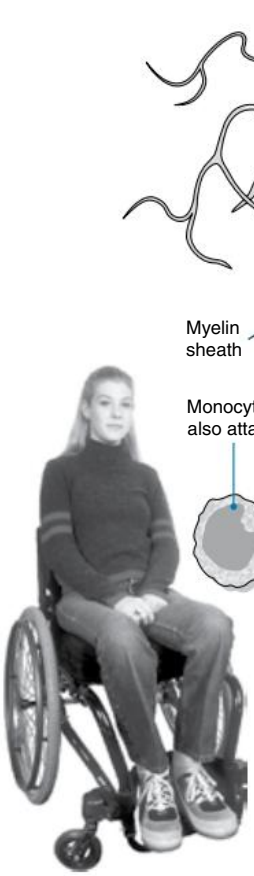
106 Autoimmune Diseases

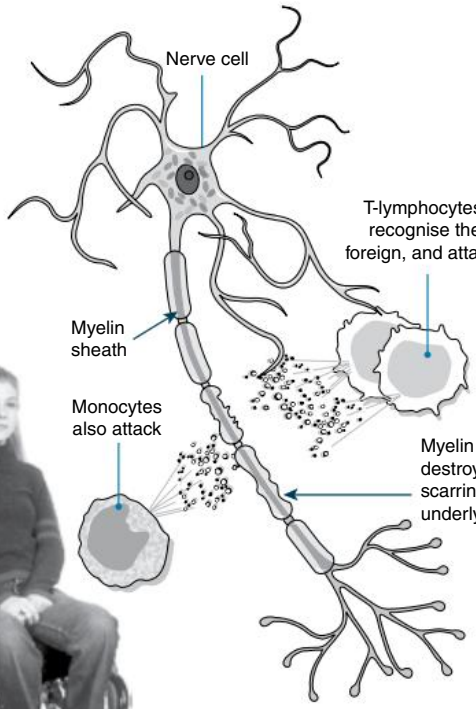
Key Idea: Autoimmune diseases are caused when the body's immune system begins to attack the body's own tissues.

Any of numerous disorders, including rheumatoid arthritis, type 1 diabetes mellitus, and multiple sclerosis, are caused by an immune system reaction to the body's own tissues. The immune system normally distinguishes self from non-self. **Autoimmune diseases** occur when this normal recognition system fails and a certain cell or tissue type is no longer recognised as self. The exact mechanisms behind autoimmune malfunctions are not fully understood, but pathogens or drugs may play a role in triggering an autoimmune response in someone who already has a genetic predisposition. The reactions are similar to those that occur in allergies, except that in autoimmune disorders, the hypersensitivity response is to the body itself, rather than to an outside substance.

Multiple Sclerosis

Multiple sclerosis (MS) is a progressive inflammatory disease of the central nervous system in which scattered patches of myelin (white matter) in the brain and spinal cord are destroyed. Myelin is the fatty connective tissue sheath surrounding conducting axons and its destruction results in the symptoms of MS: numbness, tingling, muscle weakness and paralysis.





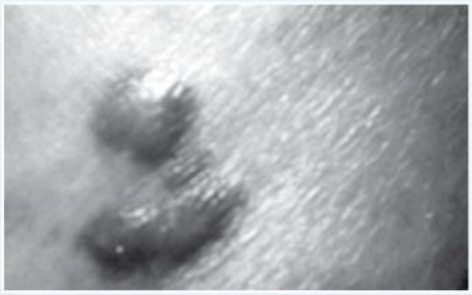
MS usually starts early in adult life and the disease is characterised by a patchy pattern of disabilities, often with dramatic unpredictable improvements. There is a genetic component to the disease, as relatives of affected people are eight times more likely to contract the disease.

Other Immune System Disorders



UCSD School of Medicine: Charles Goldberg

Rheumatoid arthritis is an auto immune disease that causes joint inflammation, usually in the hands and feet, and results in destruction of cartilage and painful, swollen joints. The disease often begins in adulthood, but can also occur in children or the elderly. Rheumatoid arthritis affects more women than men and is treated with anti-inflammatory and immunosuppressant drugs, and physiotherapy.



CDC

Lacking a sufficient immune response is called immune deficiency, and may be either congenital (present at birth) or acquired as a result of drugs, cancer, or infectious agents (e.g. HIV infection). HIV causes AIDS, which results in a steady destruction of the immune system. Sufferers then succumb to opportunistic infections and rare cancers such as Kaposi's sarcoma (above).

1. Explain the basis of the following autoimmune diseases:

   (a) Multiple sclerosis: _____

   _____

   (b) Rheumatoid arthritis: _____

   _____

2. Why are autoimmune diseases difficult to treat effectively? _____

   _____

   _____

3. Why do sufferers of immune deficiencies, such as AIDS, develop a range of debilitating infections? _____

   _____

   _____

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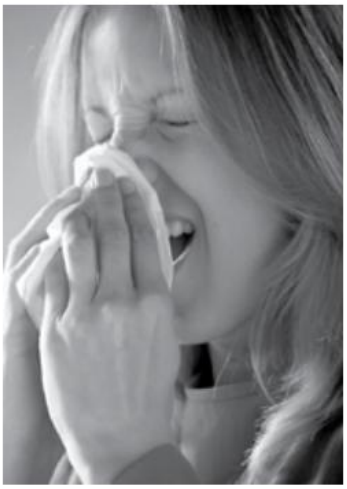
107 Allergies and Hypersensitivity

Key Idea: Hypersensitivity occurs when the immune system overreacts to an antigen or reacts to the wrong substance. Histamine plays a significant role in this response.

Sometimes the immune system may overreact, or react to the wrong substances instead of responding appropriately. This is termed **hypersensitivity** and the response leads to tissue damage rather than immunity. Hypersensitivity reactions occur after a person has been **sensitised** to an antigen. In some cases, this causes only minor discomfort (e.g hayfever), but in extreme cases it can cause death.

Hypersensitivity

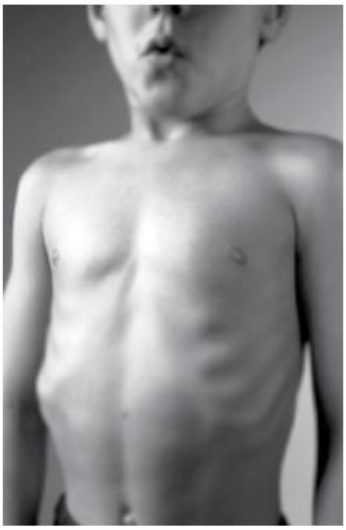
Hay fever (allergic rhinitis) is an allergic reaction to airborne substances such as dust, moulds, pollens, and animal fur or feathers. Allergy to wind-borne pollen is the most common, and certain plants (e.g. ragweed and privet) are highly allergenic. A person becomes **sensitised** when they form antibodies to harmless substances in the environment such as pollen or spores (steps 1-2 right). These substances, termed **allergens**, act as antigens to induce antibody production and an allergic response. Once a person is sensitised, the antibodies respond to further encounters with the allergen by causing the release of **histamine** from mast cells (step 4). It is histamine that mediates the symptoms of hypersensitivity reactions such as hay fever and asthma. These symptoms include wheezing and airway constriction, inflammation, itching and watering of the eyes and nose, and/or sneezing.



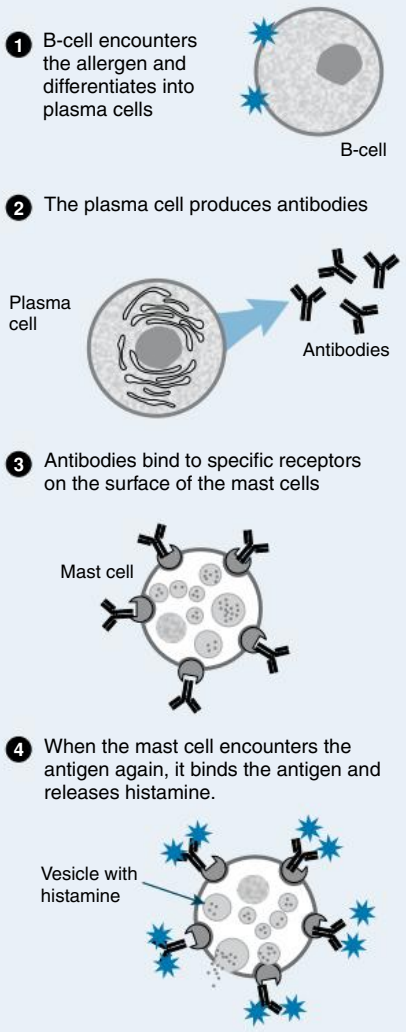
Eyewire

Asthma

Asthma is a common disease affecting 7-8% of Australian children. It usually occurs as a result of an allergic reaction to allergens such as animal dander, pollen, and the faeces of house dust mites. Asthma is a hypersensitivity reaction and involves the production of histamines from mast cells (right). The site of the reaction is the respiratory bronchioles where the histamine causes airway constriction, accumulation of fluid and mucus, and inability to breathe. During an attack, breathing is laboured and the chest cavity overexpands (photo, right). Asthma treatments work by suppressing the inflammatory response to prevent attacks and dilating the airways if an attack occurs. Asthma attacks are often triggered by environmental factors such as cold air, exercise, air pollutants, and viral infections. Recent evidence has also indicated the involvement of a bacterium: *Chlamydia pneumoniae*, in about half of all cases of asthma in susceptible adults.



The Basis of Hypersensitivity



1. What is the role of histamine in hypersensitivity responses? _____

2. What happens when a person becomes **sensitised** to an allergen? _____

3. In what way is the hypersensitivity reaction a malfunction of the immune system? _____

4. (a) What do **bronchodilators** do? _____

   (b) Why are they used to treat asthma? _____

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108 Plant Defences

Key Idea: Plants possess biochemical and structural defence mechanisms, which protect them from infection and the activities of herbivorous animals that feed off them.

Some defence mechanisms are always present as part of the plant's basic make-up, while others are activated in response to an attack. **Passive defences** take the form of physical and chemical barriers. **Active defences** are produced in direct response to an infection or physical attack and act more specifically against the threat. Some plants produce chemicals that inhibit the growth others nearby (**allelopathy**).

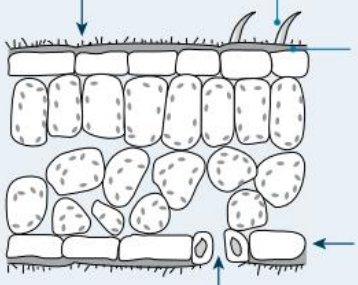
Passive Defences

Passive defences are always present and are not the result of contact with pathogens (e.g. fungi) or grazers (e.g. herbivorous mammals or insects). Passive defences may be **physical** or **chemical**. Various physical barriers help to prevent pathogens from penetrating the plant tissues. Despite these barriers, some pathogens may still gain entry into the host. If this occurs, chemical defences or more active cellular defence mechanisms (right) are used to protect the plant against further damage.

Examples of physical barriers

Hairs on the leaf surface may deter pathogens.

Thorns or **spines** on the plant surface may deter grazers.



A thick, **waxy cuticle** forms a barrier against degrading enzymes used by pathogens to gain entry to the host.

Thickened **cell walls** reduce the ability of a pathogen to invade a plant.

Stomata can be a point of entry for some pathogens. Plants may use hairs to guard these openings, or the stomata may be small enough to exclude some larger pathogens.

Examples of chemical barriers

Plants can cover their surfaces with compounds that **inhibit** the development of **pathogens**.

Many plants have developed distasteful chemicals as a defence to deter insects and other grazers.

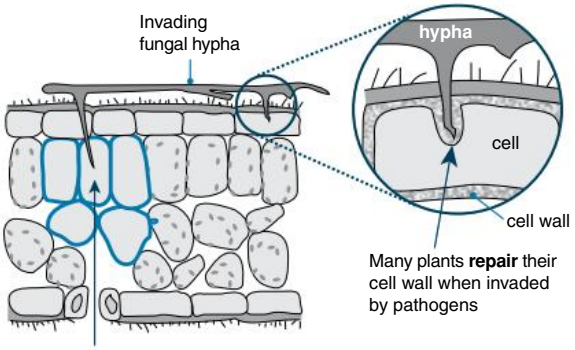


Bracken (*Pteridium aquilinum*) contains hormones that disrupt the development of insect predators.

Bracken also contains a powerful carcinogen that is toxic to livestock when eaten.

Active Defences

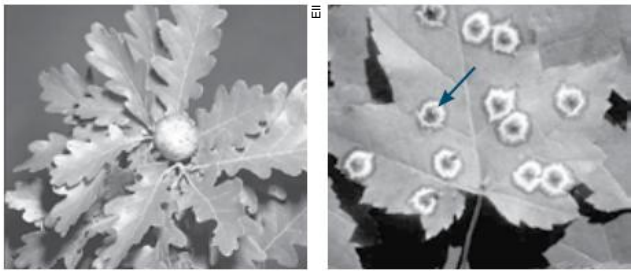
Once infected, a plant needs to respond actively to prevent any further damage. **Active defences** are invoked only after the **pathogen** has been recognised, or after wounding or attack by a herbivore. Many plant defences contribute to slowing pathogen growth without necessarily stopping it.



Many plants produce an enzyme-activated **hypersensitive response** when invaded by pathogens. This leads to the production of reactive nitric oxide and cell death. Cell death in the infected region limits the spread of the pathogen.

Other cellular active defences

Phytoalexins	Antimicrobial substances that destroy a range of pathogens, e.g. by puncturing the cell wall or disrupting metabolism.
Reactive oxygen levels	An increase in reactive oxygen species (e.g. H_2O_2) in cells kills pathogens.
Wound repair	Infected areas are sealed off by layers of thickened cells called **cork cells**.



Sealing off infected areas gives rise to abnormal swellings called galls (oak gall, above left; bulls-eye galls on a maple leaf, above right). These galls limit the spread of the parasite or the infection in the plant.

1. Distinguish between **passive** and **active** defence mechanisms: _____

2. How are **galls** effective in reducing the spread of infection in some plants? _____

3. What similarities are there between the active defence mechanisms of plants and the immune responses of animals?

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109 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its correct definition, as identified by its preceding letter code.

active immunity

antibodies (*sing.* antibody)

antigen

autoimmune disease

B cell

cell mediated immunity

clonal selection

fever

humoral immunity

immunity

immunological memory

infection

inflammation

interferon

leucocytes

lymphocytes

macrophage

monoclonal antibodies

non-specific defences (innate immunity)

passive immunity

pathogen

phagocyte

primary response

secondary response

specific (=adaptive) immune response

thymus

vaccination

A A rise in body temperature above the normal range of as a result of an increase in the body temperature regulatory set-point.

B The initial response of the immune system to exposure to an antigen.

C Resistance of an organism to infection or disease.

D Immune response involving the activation of macrophages, specific T cells, and cytokines against antigens.

E A specialised organ in the immune system with a role in producing mature T cells.

F The more rapid and stronger response of the immune system to an antigen that it has encountered before.

G The delivery by of antigenic material (the vaccine) to produce immunity to a disease.

H White blood cells, including lymphocytes, and macrophages and other phagocytes.

I Lymphocytes that make antibodies against specific antigens.

J A model for how B and T cells are selected to target specific antigens invading the body.

K Specific white blood cells involved in the adaptive immune response.

L Gamma globulin proteins in the blood or other bodily fluids, which identify and neutralise foreign material, such as bacteria and viruses.

M Large white blood cells within tissues, produced by the differentiation of monocytes.

N A disease resulting from an overactive immune response against substances and tissues normally present in the body.

O Long-lasting immunity that is induced in the host itself by the antigen.

P The invasion of a host organism by a pathogen to the detriment of the host.

Q The ability of the immune system to respond rapidly in the future to antigens encountered in the past

R Generalised defence mechanisms against pathogens, e.g. physical barriers, secretions, inflammation, and phagocytosis.

S Antibodies made by one type of immune cell that are all clones of a unique parent cell.

T Immune response that is mediated by secreted antibodies.

U A disease-causing organism.

V White blood cells that destroy foreign material, e.g. bacteria, by ingesting them.

W Anti-microbial protein made and released by lymphocytes in response to the presence of pathogens.

X The protective response of vascular tissues to harmful stimuli, such as irritants, pathogens, or damaged cells.

Y A molecule that is recognised by the immune system as foreign.

Z Immunity gained by the receipt of ready-made antibodies.

AA A pathogen and antigen-specific immune response with both cell-mediated and humoral components. Characterised by immunological memory.

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110 Detecting and Responding Synoptic Question

1. Temperature regulation is important for the body to maintain homeostasis. Discuss the case of fever in relation to homeostasis and explain its role as part of the immune response.

2. Discuss how the immune system responds to the presence of a pathogen and how this initial exposure can lead to a quicker response following a second exposure. Explain how this phenomenon is used in vaccination against disease.

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QLD As required
VCE Unit 4 (1)
WA ATAR Unit 3
SA Theme M, O

DNA and the Genetic Code

Key terms

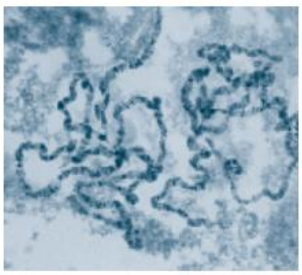
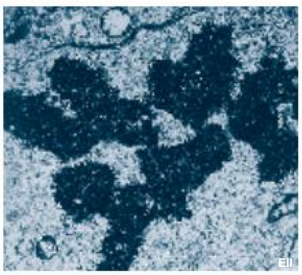
allele
anticodon
autosome
base-pairing rule
chromatid
chromosome
coding strand (mRNA)
diploid
DNA
double-helix
exons
gene
gene expression
genetic code
genome
haploid
histone
homologue
hydrogen bonding
introns
karyotype
nucleic acids
nucleotides
protein
purine
pyrimidine
ribosome
sex chromosome
RNA (mRNA, rRNA, tRNA)
template strand (mRNA)
transcription
translation

DNA and chromosomes

Learning aims and skills

Activity number

- ☐ 1 Explain what is meant by the genome of an organism and compare genome sizes in organisms from different taxonomic groups. — 111 152
- ☐ 2 Describe the nature of the bacterial chromosome and extra-chromosomal plasmids. Explain why bacteria are haploid for most genes. — 112 113
- ☐ 3 Describe the structure of eukaryote chromosomes, explaining how DNA is packaged and organised in the nucleus by histones. — 114
- ☐ 4 Explain what is meant by a karyotype and distinguish it from a karyogram. Outline the procedure for producing karyograms and describe their applications. — 115
- ☐ 5 Demonstrate an understanding of the terms homologous chromosome, chromatid, centromere, autosome, and sex chromosome by analysing karyograms. Distinguish between the diploid (2N) and haploid (1N) state of a cell. — 116
- ☐ 6 Recall the structure of DNA and explain the evidence for it. In what way did the structure of DNA provide a mechanism for its self-replication? — 11 117
- ☐ 7 Distinguish between exons and introns in DNA and describe their significance. — 117
- ☐ 8 Describe the Watson-Crick double-helix model of DNA. Include reference to the base-pairing rule, the anti-parallel strands, and the role of hydrogen bonding between purines and pyrimidines. — 11 118

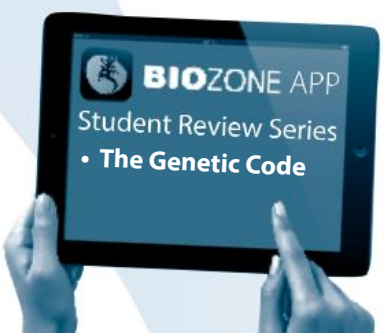




The genetic code

Learning aims and skills

Activity number

- ☐ 9 Recall the structure of RNA and describe the range of RNA molecules and their roles in cellular activities. — 119
- ☐ 10 Describe the experimental evidence that DNA was the genetic information. — 120
- ☐ 11 Explain the features of the genetic code, including: — 121 122
  - The 4-letter alphabet and the 3-letter triplet code (codon) of base sequences.
  - The non-overlapping, linear nature of the code, which is read from start to finish in one direction. The specific punctuation codons and their significance.
  - The universal nature and degeneracy of the code.
- ☐ 12 Explain what is meant by a gene. Recognise transcription and translation as the two stages of gene expression. Describe the simple one gene-one polypeptide model for gene expression. — 122 126
- ☐ 13 Describe the formation of mRNA in transcription, including the role of RNA polymerase and the significance of the coding (sense) strand and template (antisense) strand. — 123
- ☐ 14 Explain how the one gene-one polypeptide model has been modified in light of our current understanding of introns and exons in eukaryotic DNA. Describe the post-transcriptional modification of mRNA, explaining how alternative splicing of mRNA increases the number of proteins an organism can make. — 124
- ☐ 15 Describe translation (protein synthesis), including the role of tRNA, anticodons, and the general structure and role of ribosomes. — 125



111 Genomes

Key Idea: A genome is the entire, unique haploid complement of genetic material of a cell or organism.

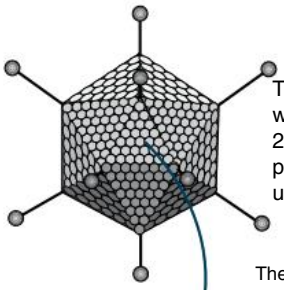
Each species has a unique genome, although there is a small amount of genetic variation between individuals within a species. For example, in humans the average genetic difference is one in every 500-1000 bases. Every cell in an individual has a complete copy of the genome. The base sequence below is the total DNA sequence for the genome of a virus. There are nine genes in the sequence, coding for nine different proteins. At least 2000 times this amount of DNA would be found in a single bacterial cell. Half a million times the quantity of DNA would be found in the genome of a single human cell. The first gene has been highlighted blue, while the start and stop codes are in dark blue rectangles.

Genome for the ϕX174 bacterial virus

Start

The blue area represents the nucleotide sequence for a single gene

CCGTCAGGATTGACACCCTCCCAATTGTATGTTTTCATGCCTCCAAATCTTGGAGGCTTTTATGGTTCGTTCTTATTACCCTTCTGAATGTCACGCTG
ACGAATACCTTCGGTTCGTAACCCCTAACTCTTTCTCATCTTTACGGTGTTCGGAGTTATCGTCCAAATTCTGGGAGCTATGGGAGTTTCAGTTTTATTA
GATGGATAACCGCATCAAGCTCTTGGAAGAGATTCTGTCTTTTCGTATGCAGGCCCTTGAGTTCGATAATGGTGATATGTATGTTGACGGCCATAAGGCT
ACAATAATTATAGTTCAACCCCCTCGTGTAACATCGTAACACGGTTAAGTAGGTAATTGAAGAGTCATTGTCTATGTTTGAGTAGTGCTTGGAGTCTTGG
CTATAGACCACCGCCCCGAAGGGGACGAAAAATGGTTTTTACAGAACGAGAAGACGGTTACGCAGTTTTGCCGCAAGCTGGCTGGTGAACGCCCTCTTAA
TTTCGGACATGCGCTATAGAATCAGGTCCGGACCTCGTTAGAACTTGTGAGTAGGAATTATGGAAAGAAAAACCCCATTAATATGAGTAGCGCTTATAGG
GCTATTCAGCGTTTGATGAATGCAATGCGACAGGCTCATGCTGATGGTTGGTTTATCGTTTTTGACAGTCTCACGTTGGCTGACGACCGATTAGAGGCGT
GTGAGGCGCACAGTTAGTAATCGGAACGCTCGGAGCCGTGGTTCTTGGTATGCTGGTTATAGTGCTTTTATCAGTGCGTTTCGTAACCCTAATAGTATTT
GTATCAGTATTTTTGTGTGGCTGAGTATCGTAGAGCTAATGGCCGTCTTCATTTCCATGCGGTGCACTTTATGGGGACACTTCGTAGAGGTAGCGTTGAG
CGCACATGGGTTGAGGCTACCCGTATGAGATTGGTATTCCGGTGCATAAAAGGTTCGATAAATTGAGCCGCGGTAACGCATAGGCTGCTGGTTTTAATCG
AGGACGCTTTTTCACGTTGTCGTTGGTTGTGGCCTGTTGATGCTAAAGGTGAGGCGCTTAAAGGTACCAGTTATATGGCTGTTGGTTTCTATGTGGCTAA
GAAGGCTTCATGGGTGTCGAACCAAAAATCAGTCAACAAGGTAAGAAATGGAACAACTCGAAATGGTCGTTCCAGGTATAGAGTGAAAAAGAATTGCATA
AAGGTGTTCAGAATCAGAATGAGCCGCAAGTTCGGGATGAAAATGGTCACAATGAGAAATCTCTCCACGGAGTGGTTAATCGAAGTTAGCAAGGTGGGTT
ACGCGGCGGTTTTGCAGCCGATGTCATTGAAAAGGGTCGGAGTTAGAGTACAGAGAAAAAGGCAAGACGAAGTTATAGACCAACTTGCCGCAGCGCAGCA
AGCTGTGACGAGAAATCTGGTCAAATTTATGCGCGCTTGGATAAAAATGATTGGCGTATCCAACCTGGAGAGTTTTATCGGTTCCATGAGGCAGAAGTTA
AAGGGGGTCGTCAGGTGAAGGTAAATTAAGCATTTGTTCGTCATCATTAAGGACGAAATAGTTCTATTAAAAAGGTGAGTAGTCTTTATAGGCTTTCACA
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AATAAAGGATGTGTTTAATCTCGGTTATGGTAGTGGAAATGGGAGAAAGGTCTTTAACAAGGTTCATAGCCGTTG



ϕX174 bacteriophage

This virus consists of DNA packaged within a protein coat made up of a 20-sided polyhedron. Spikes made of protein at each of the 12 corners are used to attach itself to a bacterial cell.

The entire DNA sequence for the virus is made up of just 9 genes

1. Explain what is meant by the **genome** of an organism: _____

_____

2. Determine the number of bases, kilobases, and megabases in this genome (100 bases in each row, except the last):

   1 kb = 1 kilobase = 1000 bases **1 Mb** = 1 megabase = 1,000,000 bases

   (a) Bases: _____ (b) Kilobases: _____ (c) Megabases: _____

3. Determine how many bases are present in the gene shown above (in the blue area): _____

4. Human DNA has around 22,500 genes. Calculate many times more genes there are in a human than in the ϕX174 virus:

_____

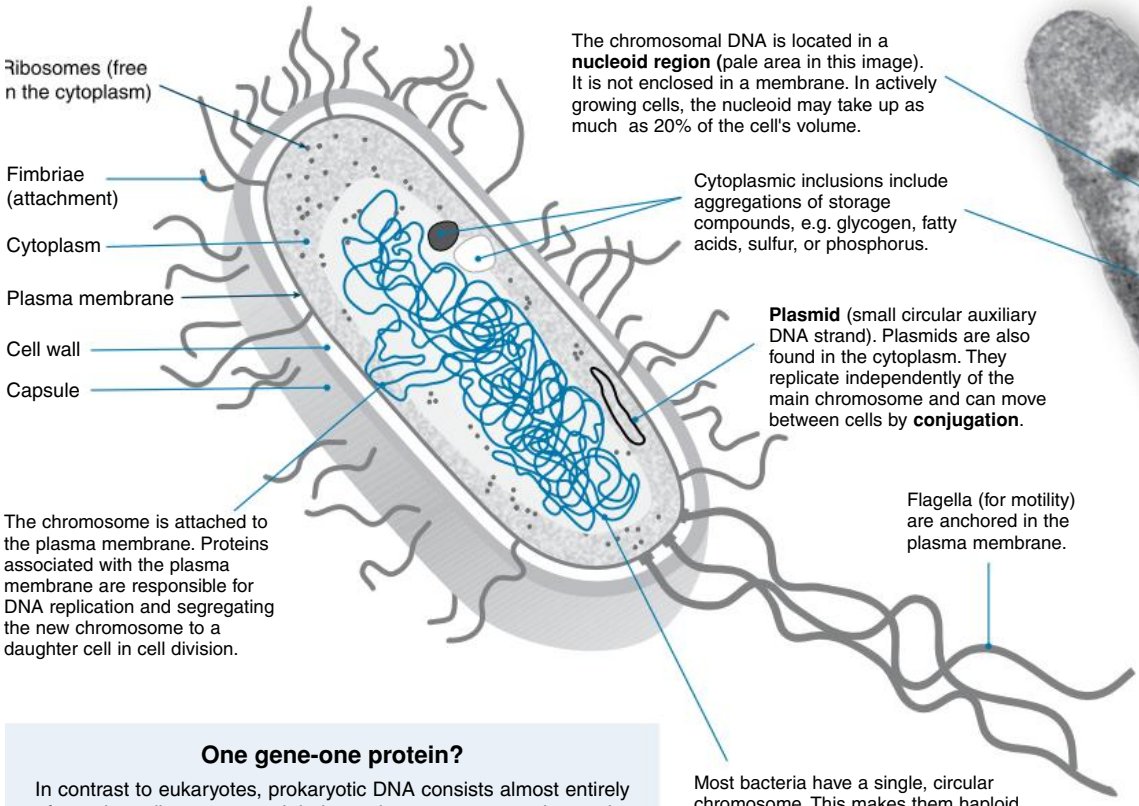
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112 Prokaryotic Chromosomes

Key Idea: Prokaryote DNA is packaged as one single chromosome that is not associated with protein.

DNA is a universal carrier of genetic information but it is packaged differently in prokaryotic and eukaryotic cells. Unlike eukaryotic chromosomes, the prokaryotic chromosome is not enclosed in a nuclear membrane and is not associated with protein. It is a single circular (rather than linear) molecule of double stranded DNA, attached to the plasma membrane and located in a nucleoid region, which is in direct contact with the cytoplasm. As well as the bacterial chromosome, bacteria often contain small circular, double-stranded DNA molecules called plasmids. Plasmids are independent of the main bacterial chromosome and usually contain 5-100 genes that are not crucial to cell survival under normal conditions.

Organisation of the Prokaryotic Chromosome



One gene-one protein?

In contrast to eukaryotes, prokaryotic DNA consists almost entirely of protein coding genes and their regulatory sequences. It was the study of prokaryotic genomes that gave rise to the one gene-one protein hypothesis, which still largely holds true for bacteria.

1. Describe three important ways in which prokaryote and eukaryote chromosomes differ:

   (a) _____

   _____

   (b) _____

   _____

   (c) _____

   _____

2. Explain the consequences to protein synthesis of the prokaryotic chromosome being free in the cytoplasm:

   _____

   _____

3. Most of the bacterial genome comprises protein coding genes and their regulatory sequences. What is the consequence of this to the relative sizes of bacterial and eukaryotic chromosomes:

   _____

   _____

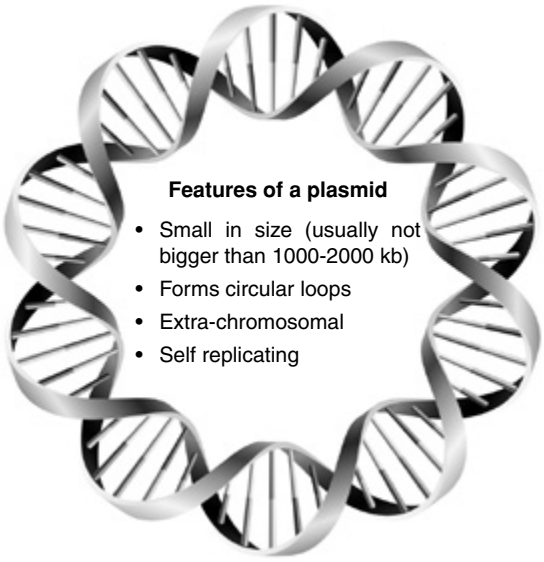
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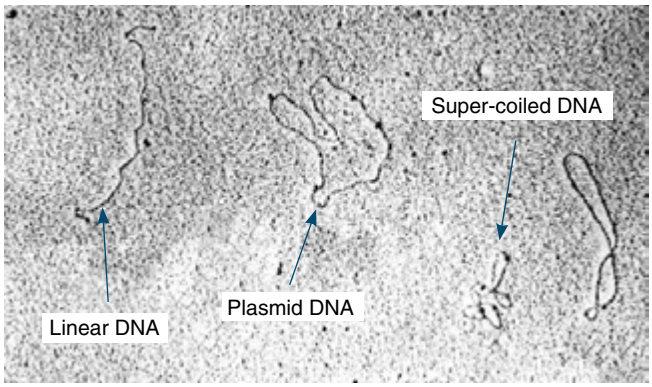
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113 Plasmid DNA

Key Idea: Plasmids are small circular pieces of DNA that are not associated with the chromosomal DNA.

Prokaryotes store most of their genetic information in one large chromosome. However, a small percentage can be found as independently replicating, circular, extra-chromosomal pieces of DNA known as **plasmids**. Plasmids may carry important genes, such as those for the production of toxins that eliminate prokaryote competitors. Plasmids are less common in eukaryotes but some species, such as the yeast *Saccharomyces,* do have them. The genetic material from viruses may form plasmid-like structures called episomes once they have infected a cell.

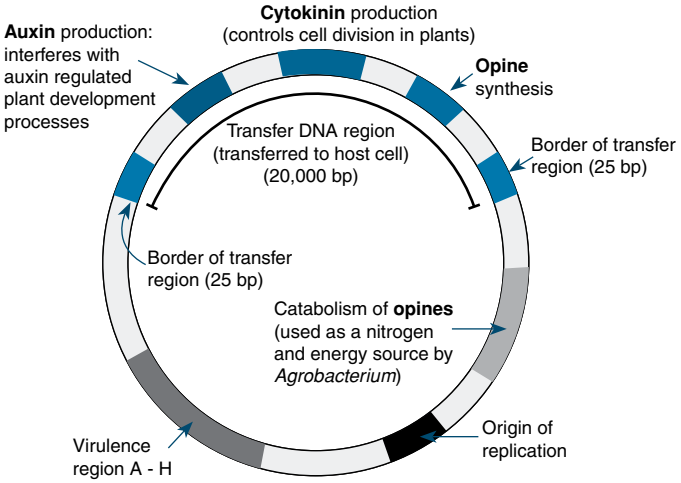




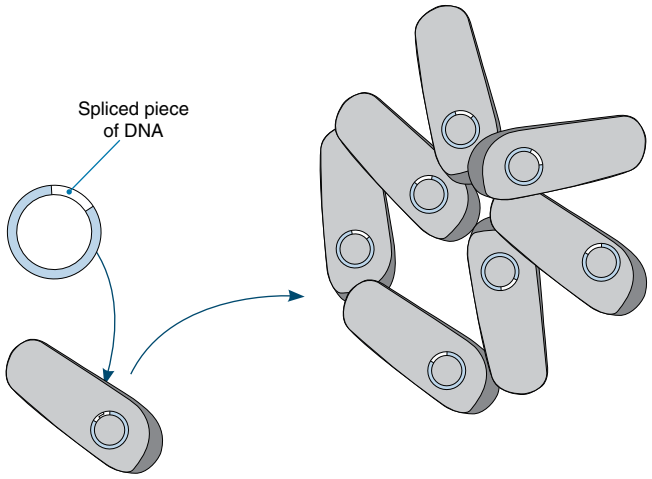
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Plasmids vary in size from 1000 base pairs (bp) to hundreds of thousands of base pairs. In bacteria, they play an important role in providing extra genetic material that confers properties such as antibiotic resistance.

Plasmids can be transferred between bacterial cells by a process of plasmid transfer called **conjugation**. Conjugation enables bacteria to obtain genetic material from other individuals by **horizontal gene transfer** (transfer of genetic material to individuals other than offspring) and it is an important mechanism for the spread of antibiotic resistance.



The bacterium *Agrobacterium tumefaciens* often contains the T*i* (tumour inducing) plasmid. This plasmid is able to transfer genetic material into plant cells and causes crown gall disease. Several regions on the plasmid (identified above) help it to infect plants. The plasmid is just over 200,000 bp long and contains 196 genes. The mapping of its genes has made it of great importance in the creation of transgenic plants.



Plasmids have provided a tool with which to introduce novel genetic material into an organism. A new gene can be spliced into a plasmid and the plasmid inserted into a recipient organism (e.g. a bacterium). The bacteria will then produce the product encoded by the gene. This methodology has enabled the industrial-scale production of valuable gene products, such as human insulin, from genetically engineered organisms.

1. What is a plasmid? _____

_____

2. Explain how a plasmid can convey a survival advantage to bacteria under certain conditions: _____

_____

_____

3. (a) Why are plasmids (such as the T*i* plasmid) useful to genetic engineers? _____

_____

(b) Into which region of the T*i* plasmid would you insert a gene in order for it to be transferred into a host plant cell?

_____

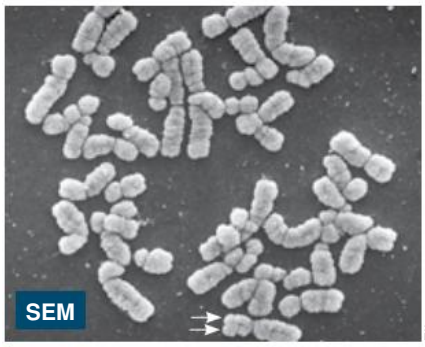
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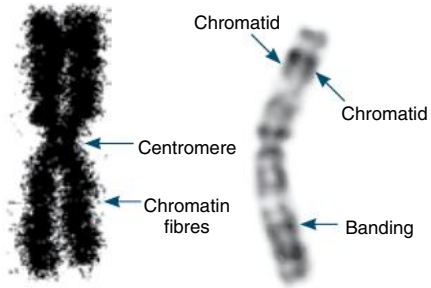
114 Eukaryote Chromosome Structure

Key Idea: A chromosome consists of DNA complexed with proteins to form a highly organised, tightly coiled structure.

The DNA in eukaryotes is complexed with histone proteins to form chromatin. The histones assist in packaging the DNA efficiently so that it can fit into the nucleus. Prior to cell division, the chromatin is at its most compact, forming the condensed metaphase chromosomes that can be seen with a light microscope.

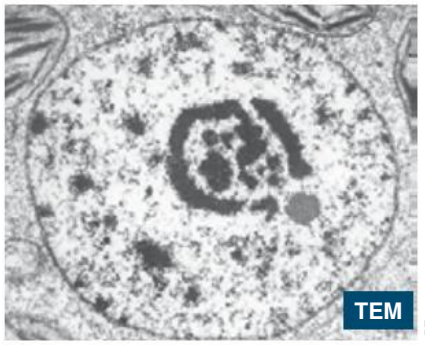


A cluster of human chromosomes seen during metaphase of cell division. Individual chromatids (arrowed) are difficult to discern on these double chromatid chromosomes.



Chromosome TEM Human chromosome 3

A human chromosome from a dividing white blood cell (above left). Note the compact organisation of the chromatin in the two chromatids. The LM photograph (above right) shows the banding visible on human chromosome 3.



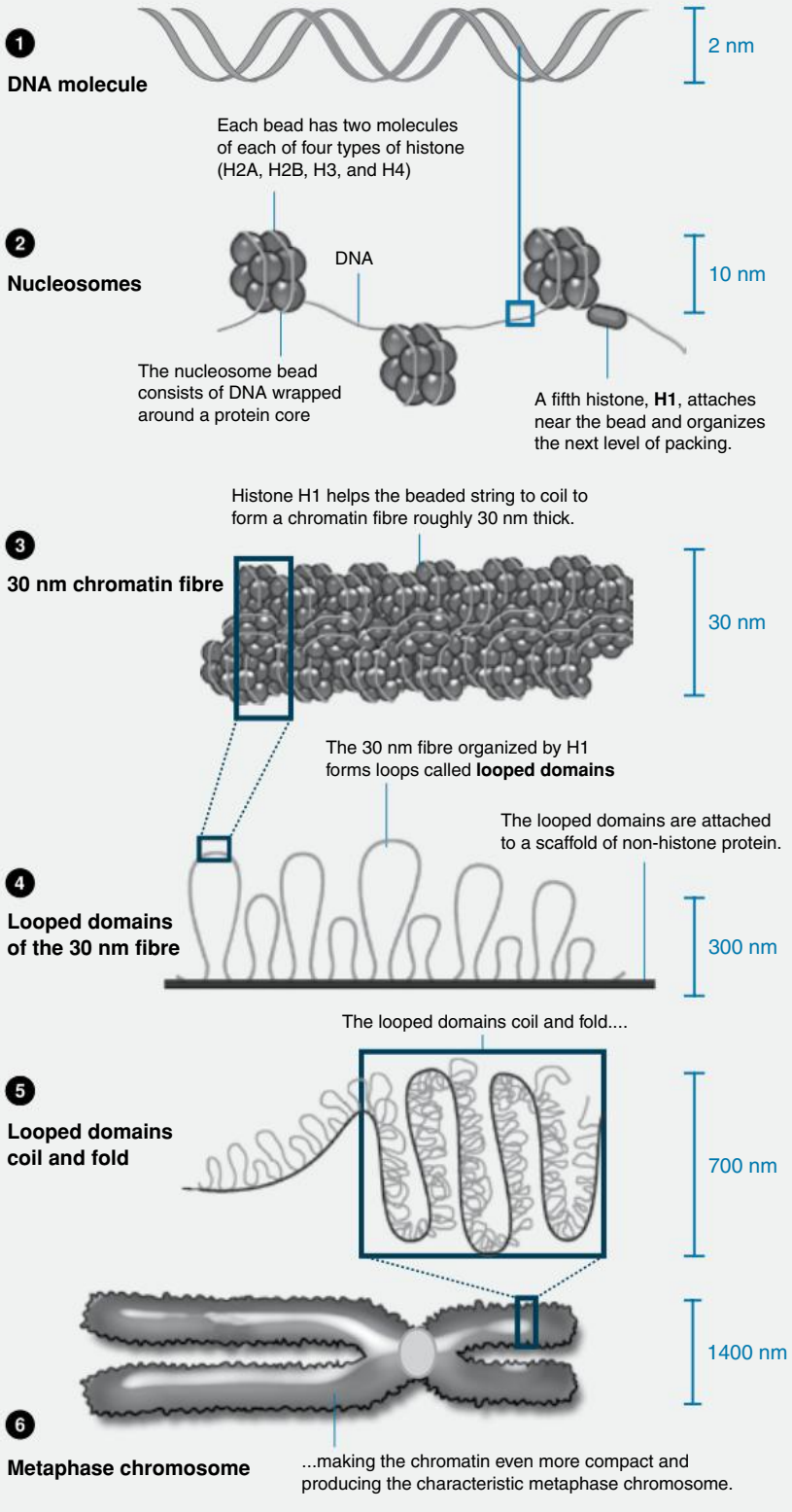
In non-dividing cells, chromosomes exist as single-armed structures. They are not visible as coiled structures, but are 'unwound' to make the genes accessible for transcription (above).



The evidence for the existence of looped domains comes from the study of giant lampbrush chromosomes in amphibian oocytes (above). Under electron microscopy, the lateral loops of the DNA-protein complex appear brushlike.

The Packaging of Chromatin

Chromatin structure is based on successive levels of DNA packing. **Histone proteins** are responsible for packing the DNA into a compact form. Without them, the DNA could not fit into the nucleus. Five types of histone proteins form a complex with DNA, in a way that resembles "beads on a string". These beads, or **nucleosomes**, form the basic unit of DNA packing.

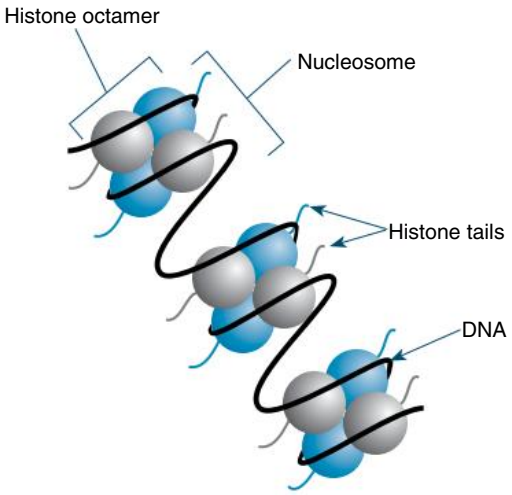


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Modifying DNA Packaging

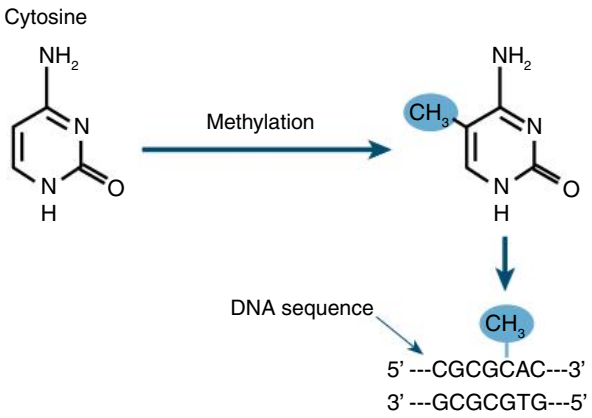
The packaging of DNA affects when (and if) genes are expressed either by making the nucleosomes in the chromatin pack together tightly (**heterochromatin**) or more loosely (**euchromatin**). This affects whether or not RNA polymerase can attach to the DNA and transcribe the DNA to mRNA. Packaging of DNA is affected by histone modification and DNA methylation.

Histone Modification



Histones may be modified by a number of processes, including methylation of the histone tail. Depending on the type of modification, the chromatin may pack together more tightly or more loosely, affecting the cell's ability to transcribe genes.

DNA Methylation



Cytosine methylation is an important process in DNA packaging and gene expression. Cytosine methylation affects gene expression in two ways: it may physically impede the binding of transcription factors or it may cause the chromatin to bind tightly together so that genes cannot be transcribed.

1. Explain the significance of the following terms used to describe the structure of chromosomes:

   (a) DNA: _____

   _____

   (b) Chromatin: _____

   _____

   (c) Histone: _____

   _____

   (d) Nucleosome: _____

   _____

2. (a) Describe the effect of histone modification and DNA methylation on DNA packaging: _____

   _____

   _____

   (b) How does this affect transcription of the DNA?_____

   _____

3. Each human cell has about 1 metre of DNA in its nucleus. Explain how this DNA is packaged into the nucleus:

   _____

   _____

   _____

   _____

   _____

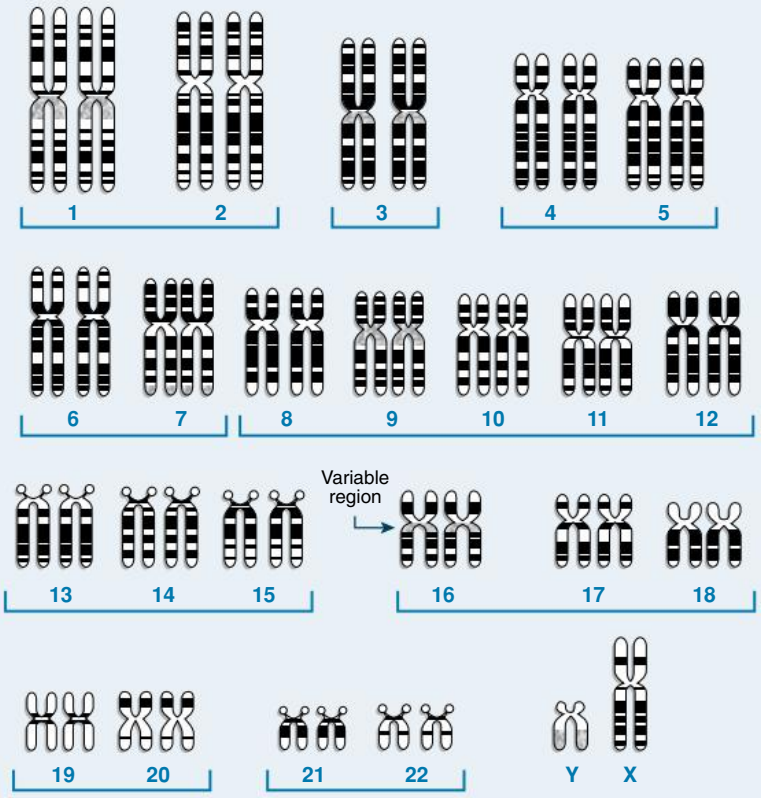
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115 Karyotypes

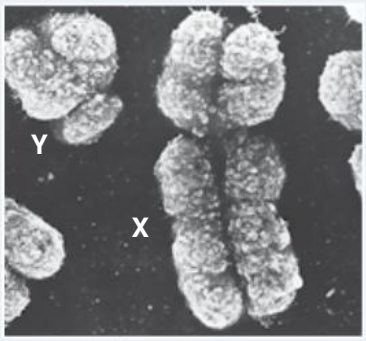
Key Idea: The karyotype is the number and appearance of chromosomes in the nucleus of a eukaryotic cell. The karyotype can be pictured in a standard format, called a karyogram, in which the chromosomes are ordered by size. The diagram below shows a **karyogram** for a normal human. Karyotyping begins with 'freezing' the nuclei of cultured white blood cells in metaphase of mitosis. A photograph of the chromosomes is then cut up and the chromosomes are rearranged on a grid to produce the karyogram. The homologous pairs are placed together, identified by their general shape, length, and banding pattern after staining. In humans, the **male karyotype** has 44 autosomes, an X chromosome, and a Y chromosome (44 + XY). The **female karyotype** has two X chromosomes (44 + XX).

Typical Layout of a Human Karyogram





A scanning electron micrograph (SEM) of human chromosomes clearly showing their double chromatids.



This SEM shows the human X and Y chromosomes. Although these two are the sex chromosomes, they are not homologous.

Karyotypes for different species

The term **karyotype** refers to the chromosome complement of a cell or a whole organism. In particular, it shows the number, size, and shape of the chromosomes as seen during metaphase of mitosis. The diagram on the left depicts the human karyotype. Chromosome numbers vary considerably among organisms and may differ markedly between closely related species:

Organism	Chromosome number (2N)
Vertebrates	
cat	38
rat	42
rabbit	44
human	46
chimpanzee	48
gorilla	48
cattle	60
horse	64
dog	78
turkey	82
goldfish	94
Invertebrates	
horse roundworm	2
fruit fly *Drosophila*	8
housefly	12
honey bee	32 or 16
Hydra	32
Plants	
broad bean	12
cabbage	18
garden pea	14
rice	24
Ponderosa pine	24
orange	18, 27 or 36
potato	48

NOTE: The number of chromosomes is not a measure of the quantity of genetic information.

1. (a) What is a **karyogram**? _____

_____

(b) What information can it provide? _____

_____

2. Distinguish between **autosomes** and **sex chromosomes**: _____

_____

_____

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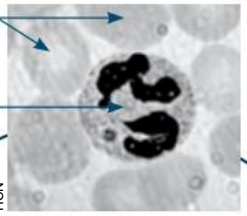
Preparing a Karyogram



1 A sample of cells is taken from the amniotic fluid around a fetus (by **amniocentesis**), from a sample from the placenta (by **chorionic villus sampling**) or from a blood sample. Amniocentesis and chorionic villus sampling both carry risks of miscarriage, so are performed after a maternal age of 35, when the chance of carrying a fetus with a chromosomal abnormality is greater than the chance of miscarriage as a result of the procedure.



2 The cells are centrifuged and treated with a solution to make the cells swell up and their chromosomes spread out.

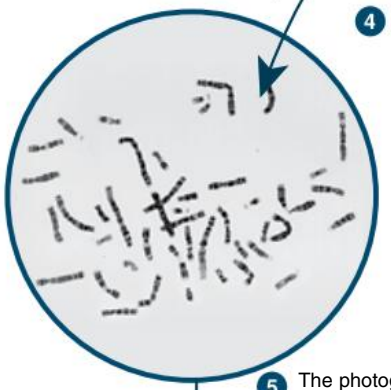


3 Lymphocytes (a type of white blood cell) are treated with a drug to cause them to go into cell division (mitosis) and are grown for several days in culture. They are treated with another drug to halt the cycle at the metaphase stage.



4 A drop of the cell suspension in preservative is spread on a microscope slide, dried and stained with a dye that causes a banding pattern to appear on each chromosome.

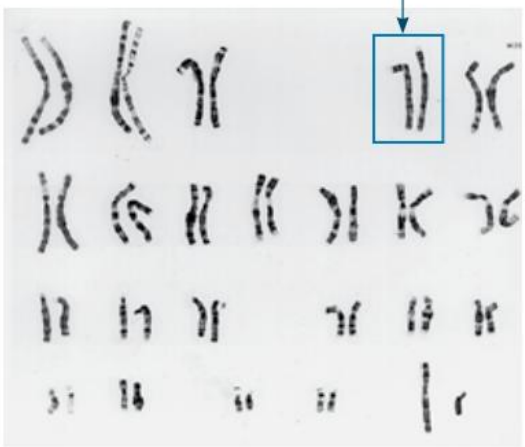




The stained white blood cells are viewed under a microscope and a clearly arranged spread of chromosomes is photographed.

5 The photograph is cut up so that each chromosome is separate from the others. This may be done manually or electronically with the aid of a computer. The chromosomes are arranged into homologous pairs according to size and shape.

Homologous pair



Karyogram: Cytogenetics Dept, Waikato Hospital

Karyogram for a male human: 44 + XY



Karyogram: Cytogenetics Dept, Waikato Hospital

Karyogram for a female human: 44 + XX

Chromosomes are arranged in groups according to size, shape and banding pattern.

3. On the male and female karyograms above, **number** each homologous pair of chromosomes using the diagram on the previous page as a guide.

4. **Circle** the sex chromosomes (**X** and **Y**) in the karyogram of the female and the male.

5. Write down the number of *autosomes* and the arrangement of *sex chromosomes* for each sex:

   (a) **Female**: No. of autosomes: _____ Sex chromosomes: _____

   (b) **Male**: No. of autosomes: _____ Sex chromosomes: _____

6. State how many chromosomes are found in a:

   (a) Normal human (somatic) body cell: _____ (b) Normal human sperm or egg cell: _____

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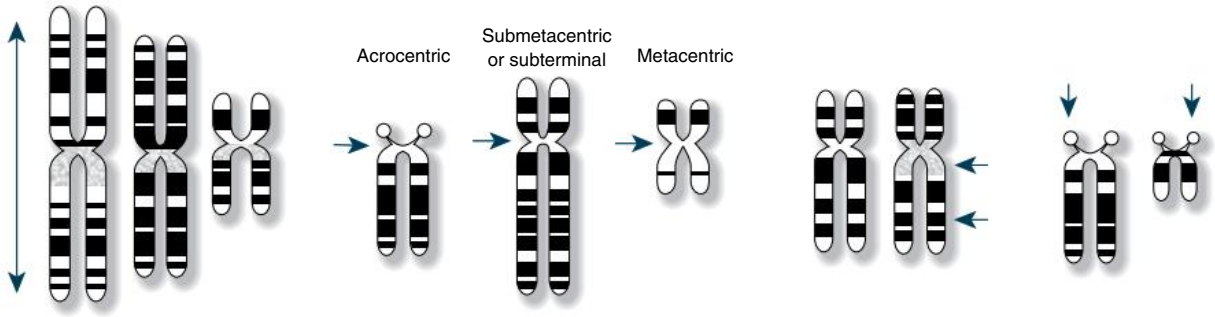
116 Human Karyotype Exercise

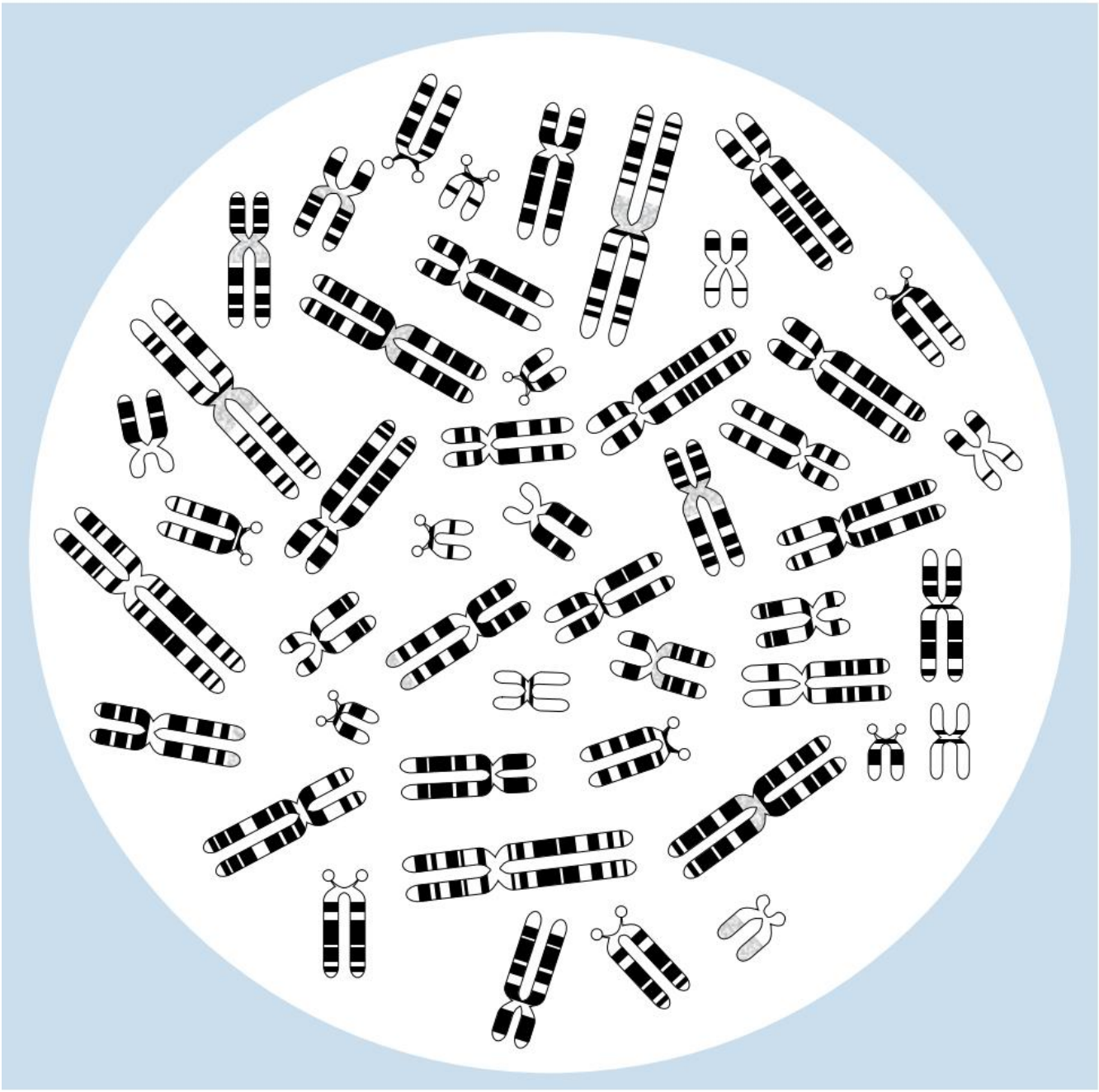
Key Idea: A karyogram can be created by matching the size and banding pattern of individual chromosomes.

Each chromosome has specific distinguishing features. Chromosomes are stained in a special technique that gives them a banded appearance in which the banding pattern represents regions containing up to many hundreds of genes.

Cut out the chromosomes below and arrange them on the record sheet in order to determine the sex and chromosome condition of the individual whose karyotype is shown. The karyograms presented on the previous pages and the hints on how to recognise chromosome pairs can be used to help you complete this activity.

Distinguishing Characteristics of Chromosomes





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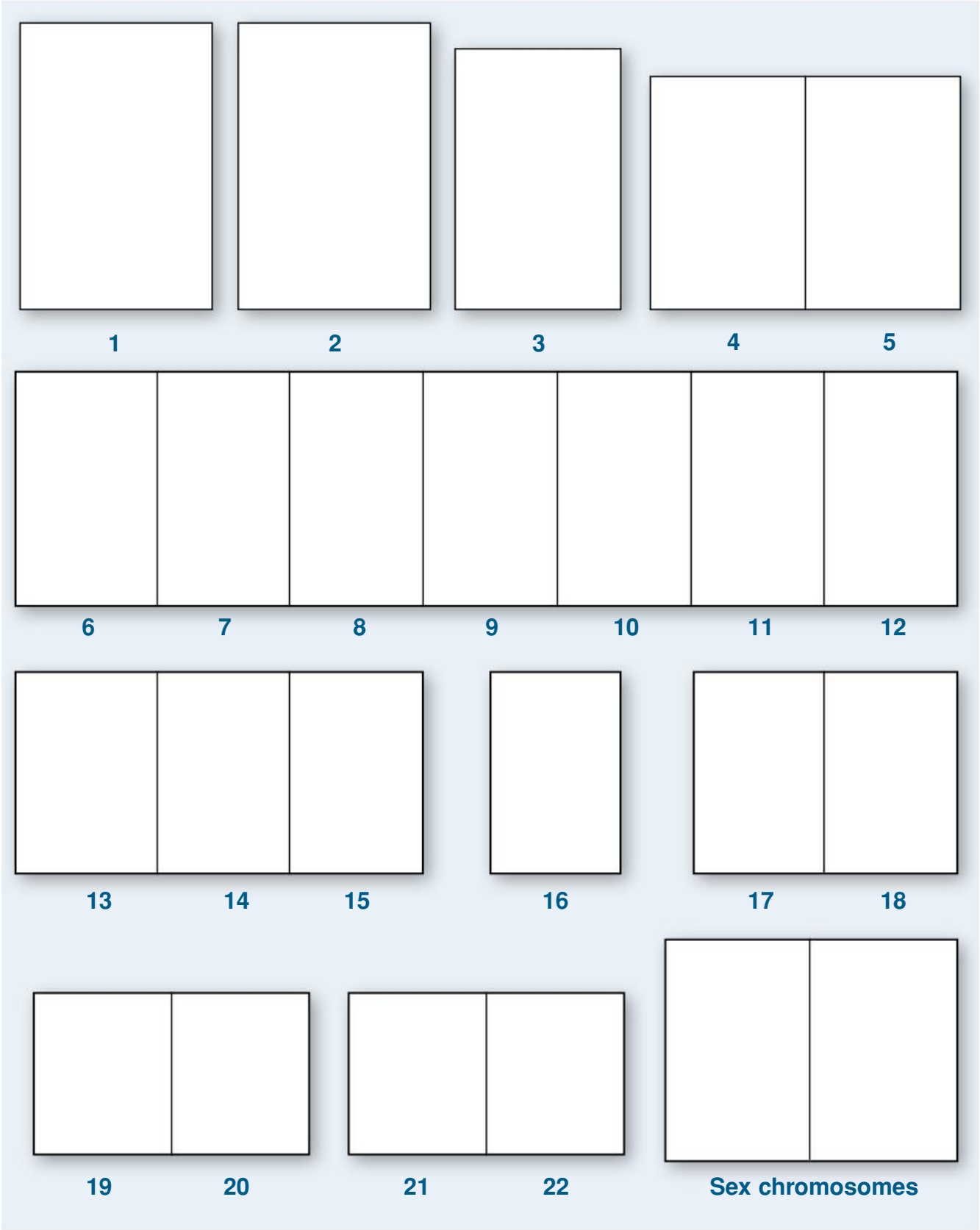
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1. Cut out the chromosomes on page 151 and arrange them on the record sheet below in their homologous pairs.
2. (a) Determine the sex of this individual: **male** or **female** (circle one)

   (b) State whether the individual's *chromosome arrangement* is: **normal** or **abnormal** (circle one)

   (c) If the arrangement is *abnormal*, state in what way and name the syndrome displayed: _____

   _____



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117 DNA Molecules

Key Idea: Once the structure of DNA was known, it immediately suggested a mechanism for its replication.

It took the work of many scientists working in different areas many years to determine the structure of DNA. The final pieces of evidence came from a photographic technique called X-ray crystallography in which X-rays are shone through crystallised molecules to produce a pattern on a film. The pattern can be used to understand the structure of the molecule. The focus of much subsequent research on DNA has been on protein-coding DNA sequences, yet protein-coding DNA accounts for less than 2% of the DNA in human chromosomes. The rest of the DNA was once called 'junk', meaning it did not code for anything. We now know that much of it codes for regulatory RNA molecules and is not junk at all.

Discovering the Structure of DNA

Although James Watson and Francis Crick are often credited with the discovery of the structure of DNA, credit must also go to at least two other scientists who were instrumental in acquiring the images that Watson and Crick were to base their discovery on. Maurice Wilkins and Rosalind Franklin produced X-ray diffraction patterns of the DNA molecule. The patterns provided measurements of different parts of the molecule and the position of different groups of atoms. Wilkins showed Franklin's X-ray image (photo 51) to Watson and Crick who then correctly interpreted the image and produced a model of the DNA molecule.

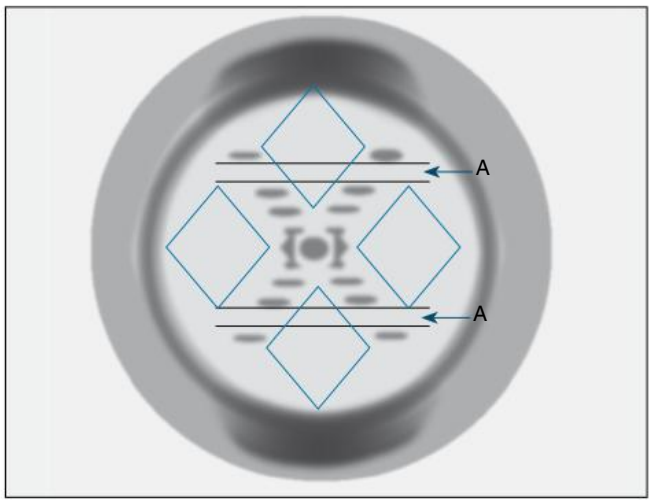
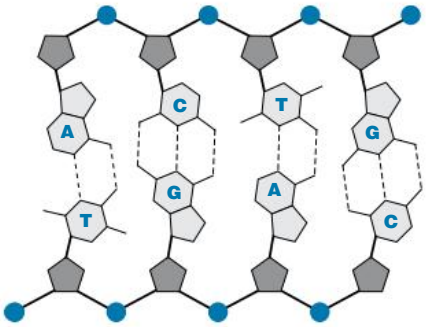


Diagram representing the image produced by Rosalind Franklin

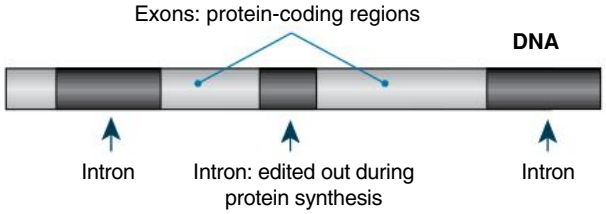
Numerous distinct parts of the X-ray image indicate specific qualities of the DNA. The distinct X pattern indicates a helix structure. However, Watson and Crick realised that the apparent gaps in the X (labelled **A**) were due to the repeating pattern of a *double* helix. The diamond shapes (shown in blue) indicate the helix is continuous and of constant dimensions and that the sugar-phosphate backbone is on the outside of the helix. The distance between the dark horizontal bands allows the calculation of the length of one full turn of the helix.

Structure and Replication



The realisation that DNA was a double helix consisting of antiparallel strands made of bases that followed a strict base pairing rule suggested a mechanism for its replication. Watson and Crick proposed that each strand served as a template and that DNA replication was semi-conservative, producing two daughter strands consisting of half new and half parent material. The proposal was confirmed by Meselson and Stahl.

Exons and Introns



Most protein-coding genes in eukaryotic DNA are not continuous. The protein-coding regions (**exons**) are interrupted by non-protein-coding regions called **introns**. Introns are edited out of the protein-coding sequence prior to translation (protein synthesis). After processing, the introns may go on to serve a regulatory function.

1. What made Watson and Crick realise that DNA was a double helix? _____

_____

_____

2. What proportion of DNA in a eukaryotic cell is used to code for proteins? _____

3. (a) Describe the organisation of protein-coding regions in eukaryotic DNA: _____

_____

_____

(b) What might be the purpose of the introns? _____

_____

_____

KNOW WEB 117 LINK 124 LINK 155

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118 Creating a DNA Model

Key Idea: Nucleotides pair together in a specific way called the base pairing rule. In DNA, adenine always pairs with thymine, and cytosine always pairs with guanine.

DNA are double stranded molecules. Each strand is made up of nucleotides. The chemical properties of each nucleotide mean it can only bind with a one other type of nucleotide. This is called the **base pairing rule**, and is explained in the table below. The exercise on the following pages will help you learn and remember this rule.

DNA Base Pairing Rule

Adenine	is always attracted to	**Thymine**	A ⟷ T
Thymine	is always attracted to	**Adenine**	T ⟷ A
Cytosine	is always attracted to	**Guanine**	C ⟷ G
Guanine	is always attracted to	**Cytosine**	G ⟷ C

1. Cut around the nucleotides on page 157 and separate each of the 24 nucleotides by cutting along the columns and rows (see arrows indicating two such cutting points). Although drawn as geometric shapes, these symbols represent chemical structures.

2. Place one of each of the four kinds of nucleotide on their correct spaces below:

Place a cut-out symbol for **thymine** here

Thymine

Place a cut-out symbol for **cytosine** here

Cytosine

Place a cut-out symbol for **adenine** here

Adenine

Place a cut-out symbol for **guanine** here

Guanine

3. Identify and **label** each of the following features on the adenine nucleotide immediately above:
   phosphate, **sugar**, **base**, **hydrogen bonds**

4. Create one strand of the DNA molecule by placing the 9 correct 'cut out' nucleotides in the labelled spaces on the following page (DNA molecule). Make sure these are the right way up (with the **P** on the left) and are aligned with the left hand edge of each box. Begin with thymine and end with guanine.

5. Create the complementary strand of DNA by using the base pairing rule above. Note that the nucleotides have to be arranged upside down.

6. Under normal circumstances, it is not possible for adenine to pair up with guanine or cytosine, nor for any other mismatches to occur. Describe the **two factors** that prevent a mismatch from occurring:

   Factor 1: _____

   _____

   Factor 2: _____

   _____

7. Once you have checked that the arrangement is correct, you may glue, paste or tape these nucleotides in place.

NOTE: There may be some value in keeping these pieces loose in order to practise the base pairing rule. For this purpose, *removable tape* would be best.

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DNA Molecule

Put the named nucleotides on the left hand side to create the template strand

Put the matching **complementary** nucleotides opposite the template strand



Thymine

Cytosine

Adenine

Adenine

Guanine

Thymine

Thymine

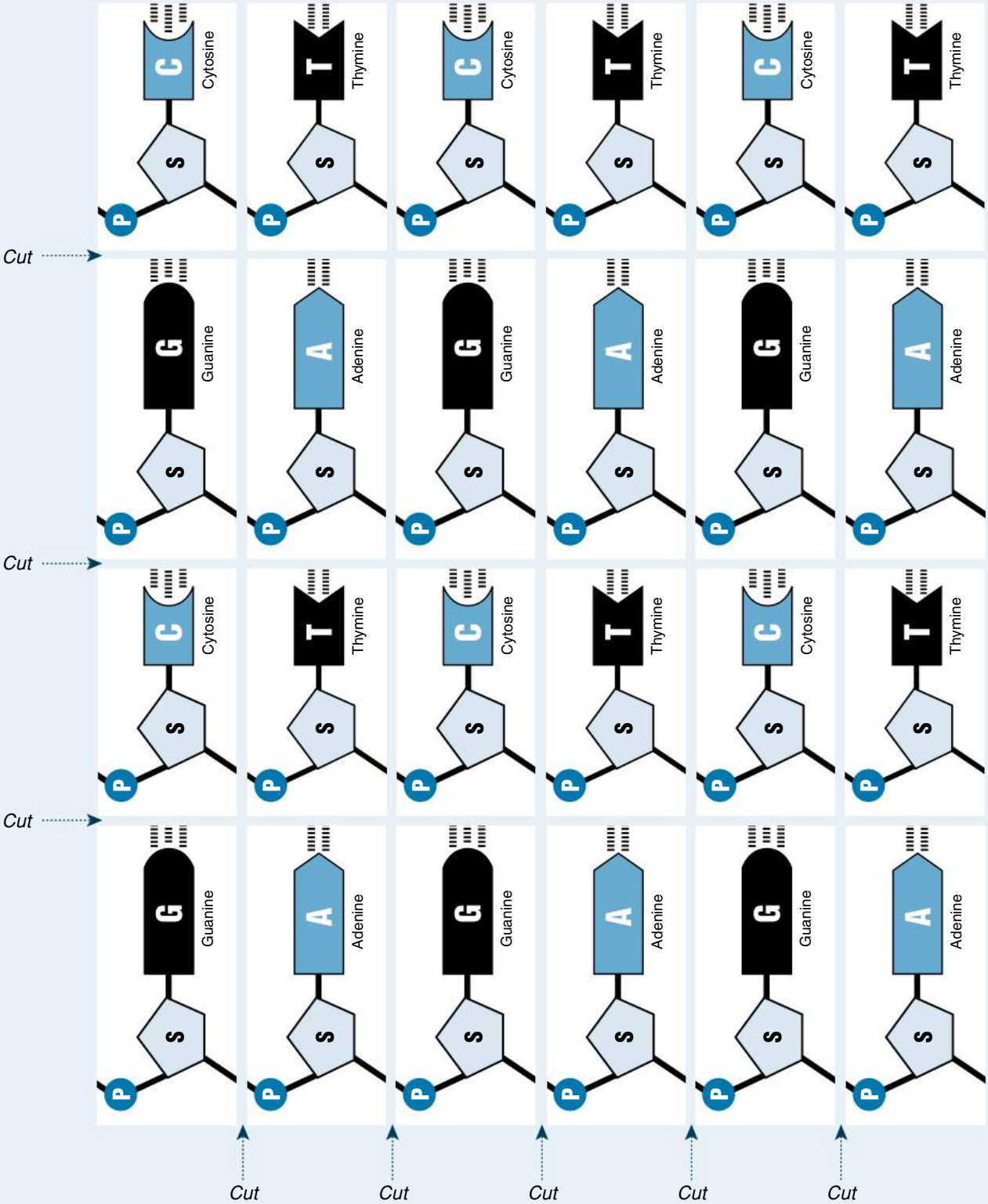
Cytosine

Guanine

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Nucleotides

Tear out this page along the perforation and separate each of the 24 nucleotides by cutting along the columns and rows (see arrows indicating the cutting points).



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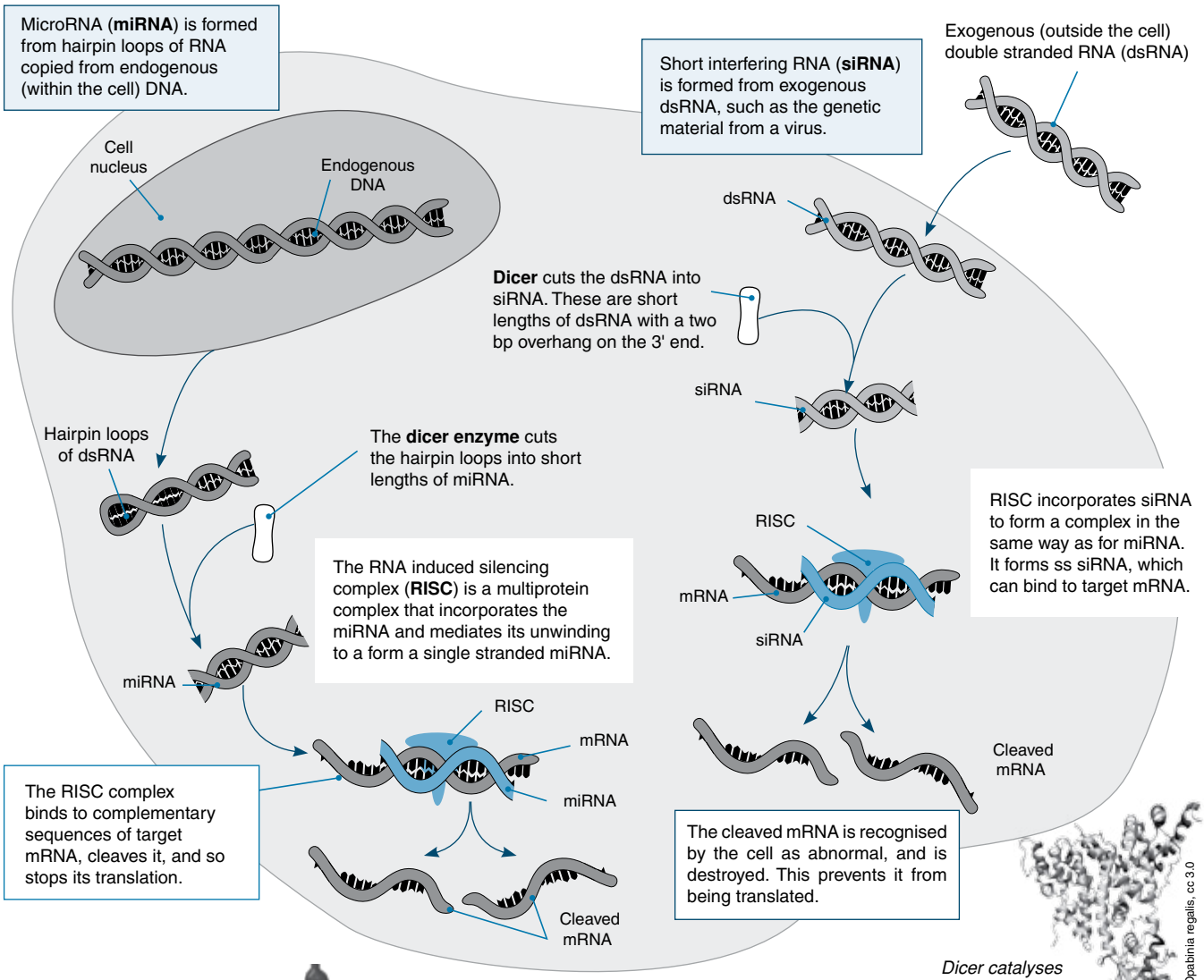
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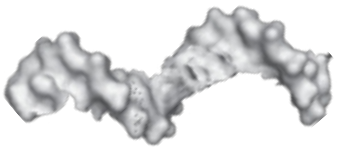
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119 RNA Molecules

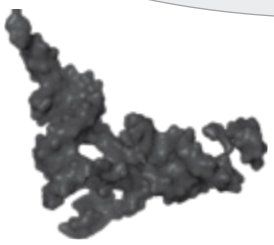
Key Idea: Different types of RNA molecules play many key roles in the regulation of gene expression.

RNA plays vital roles in transcribing and translating DNA, forming messenger RNA (mRNA), transfer RNA (tRNA), and ribosomal RNA (rRNA). RNA is also involved in modifying mRNA after transcription and regulating translation. Regulation of translation is achieved by destroying specific mRNA targets using short RNA lengths, which may be exogenous (short interfering RNAs) or endogenous (microRNAs). Mechanisms of **RNA interference** (RNAi) are illustrated below.

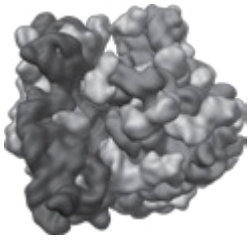




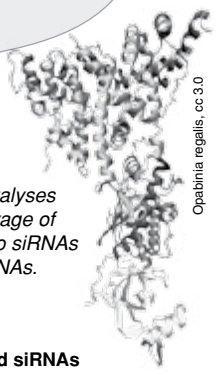
Messenger RNA (above) is transcribed from DNA. It carries a copy of the genetic instructions from the DNA to ribosomes in the cytoplasm, where it is translated into a polypeptide chain.



Transfer RNA (above) carries amino acids to the growing peptide chain. One end of the tRNA carries the genetic code in a three-nucleotide sequence called the anticodon. The amino acid links to the 3' end of the tRNA.



Ribosomal RNA (above) forms ribosomes from two separate ribosomal components (the large and small subunits) and assembles amino acids into a peptide chain.



Opabinia regalis, cc 3.0

Dicer catalyses the cleavage of dsRNA to siRNAs and miRNAs.

miRNAs and siRNAs

RNA interference (**RNAi**) regulates gene expression through miRNAs and siRNAs, which bind to specific mRNA sequences, causing them to be cleaved. RNAi is important in gene regulation during development and in defence against viruses, which often use double-stranded RNA as an infectious vector.

1. Briefly describe the roles of RNA: _____

_____

2. How does RNAi regulate gene expression? _____

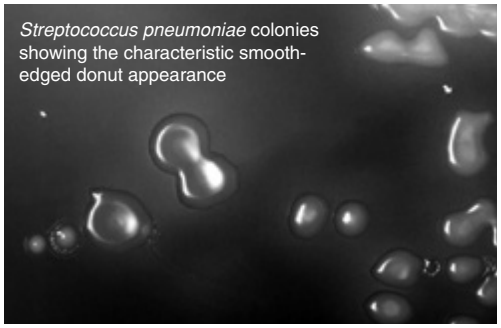
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120 DNA Carries the Code

Key Idea: A series of experiments in the 1940s and 1950s confirmed that it was DNA that carried the genetic information.



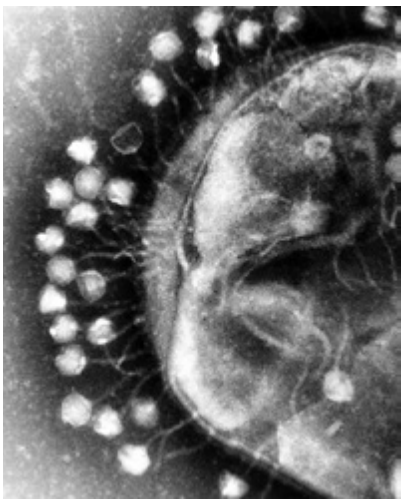
Streptococcus pneumoniae colonies showing the characteristic smooth-edged donut appearance

CDC

Scientists had known about DNA since the end of the 19th century, but its role in storing information remained unknown until the 1940s, and its structure remained a mystery for another decade after that. In 1928, experiments by British scientist Frederick Griffith gave the first indications that DNA was responsible for passing on information. Griffith had been working with two strains of the bacteria *Streptococcus pneumoniae*. Only one strain (the pathogenic strain) caused pneumonia and it was easily identified because it formed colonies with smooth edges. The other, benign strain formed colonies with rough edges. When mice were injected with the pathogenic strain they developed pneumonia and died. The mice injected with the benign strain did not. Mice injected with the heat-killed pathogenic strain did not develop pneumonia either. This showed that the disease was not caused by a chemical associated with the bacteria, or a response by the body to the bacteria, it was the bacterial cells themselves. In a second experiment, Griffith mixed the benign strain with the heat-killed pathogenic strain and injected it into healthy mice. To his surprise, the mice developed pneumonia. When bacteria from the mice were recovered and cultured they produced colonies identical to the pathogenic strain. Somehow the harmless bacteria had acquired information from the dead pathogenic strain. Griffith called this process **transformation**.



In 1944, American scientists, led by Oswald Avery, continued with Griffith's experiments. They made an extract from the heat-killed pathogenic strain and treated it with chemicals to destroy any lipids, carbohydrates, or proteins. This was mixed with the benign strain and transformation still occurred. This established that no proteins, lipids, or carbohydrates were responsible for the transformation. When another identical extract was treated with chemicals that break down DNA, the transformation did not take place - the benign strain failed to acquire the information required to cause pneumonia. From this it was deduced that DNA was the unit that was carrying the information from one bacteria to another.



Graham Colm

Another experiment in 1952 by Alfred Hershey and Martha Chase, confirmed what the other two experiments had shown. Hershey and Chase worked with viruses, which were known to have DNA and to transfer information to their host. However, there was debate over whether the information was transferred by the DNA or by the protein coat of the virus. Hershey and Chase used radioactive sulfur and radioactive phosphorus to mark different parts of the virus. The sulfur was incorporated into the protein coat while the phosphorus was incorporated into the viral DNA. The viruses were then mixed with bacteria and the infected bacteria analysed. The bacteria were found to contain radioactive phosphorus but not radioactive sulfur, showing that the virus had indeed passed information to its host by injecting its own DNA.

1. How did Griffith confirm that it was the bacteria causing the pneumonia and not something else?

2. Why were sulfur and phosphorus used in Hershey's experiment? _____

3. Why is it important to conduct two different experiments (e.g. Avery's and Hershey's) when investigating a hypothesis?

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121 The Genetic Code

Key Idea: The genetic code is the set of rules by which the genetic information in DNA or mRNA is translated into proteins. The genetic information for the assembly of amino acids is stored as three-base sequence. These three letter codes on mRNA are called **codons**. Each codon represents one of 20 amino acids used to make proteins. The code is effectively universal, being the same in all living things (with a few minor exceptions). The genetic code is summarised in a mRNA-amino acid table, which identifies the amino acid encoded by each mRNA codon. The code is degenerate, meaning there may be more than one codon for each amino acid. Most of this degeneracy is in the third nucleotide of a codon.

Amino acid	Codons that code for this amino acid	No.	Amino acid	Codons that code for this amino acid	No.
Ala Alanine	GCU, GCC, GCA, GCG	4	**Leu** Leucine		
Arg Arginine			**Lys** Lysine		
Asn Asparagine			**Met** Methionine		
Asp Aspartic acid			**Phe** Phenylalanine		
Cys Cysteine			**Pro** Proline		
Gln Glutamine			**Ser** Serine		
Glu Glutamic acid			**Thr** Threonine		
Gly Glycine			**Try** Tryptophan		
His Histidine			**Tyr** Tyrosine		
Iso Isoleucine			**Val** Valine		

1. Use the **mRNA-amino acid table** (below) to list in the table above all the **codons** that code for each of the amino acids and the number of different codons that can code for each amino acid (the first amino acid has been done for you).

2. (a) How many amino acids could be coded for if a codon consisted of just two bases?

   (b) Why is this number of bases inadequate to code for the 20 amino acids required to make proteins?

3. Describe the consequence of the degeneracy of the genetic code to the likely effect of a change to one base in a triplet:

mRNA-Amino Acid Table

How to read the table: The table on the right is used to 'decode' the genetic code as a sequence of amino acids in a polypeptide chain, from a given mRNA sequence. To work out which amino acid is coded for by a codon (triplet of bases) look for the first letter of the codon in the row label on the left hand side. Then look for the column that intersects the same row from above that matches the second base. Finally, locate the third base in the codon by looking along the row from the right hand end that matches your codon.

Example: Determine **CAG**

C on the left row,
A on the top column,
G on the right row
CAG is Gln (**glutamine**)

Read second letter here / Read first letter here → Second Letter; Read third letter here → Third Letter

First Letter	Second Letter: U	Second Letter: C	Second Letter: A	Second Letter: G	Third Letter
U	UUU Phe UUC Phe UUA Leu UUG Leu	UCU Ser UCC Ser UCA Ser UCG Ser	UAU Tyr UAC Tyr UAA STOP UAG STOP	UGU Cys UGC Cys UGA STOP UGG Trp	U C A G
C	CUU Leu CUC Leu CUA Leu CUG Leu	CCU Pro CCC Pro CCA Pro CCG Pro	CAU His CAC His CAA Gln CAG Gln	CGU Arg CGC Arg CGA Arg CGG Arg	U C A G
A	AUU Ile AUC Ile AUA Ile AUG Met	ACU Thr ACC Thr ACA Thr ACG Thr	AAU Asn AAC Asn AAA Lys AAG Lys	AGU Ser AGC Ser AGA Arg AGG Arg	U C A G
G	GUU Val GUC Val GUA Val GUG Val	GCU Ala GCC Ala GCA Ala GCG Ala	GAU Asp GAC Asp GAA Glu GAG Glu	GGU Gly GGC Gly GGA Gly GGG Gly	U C A G

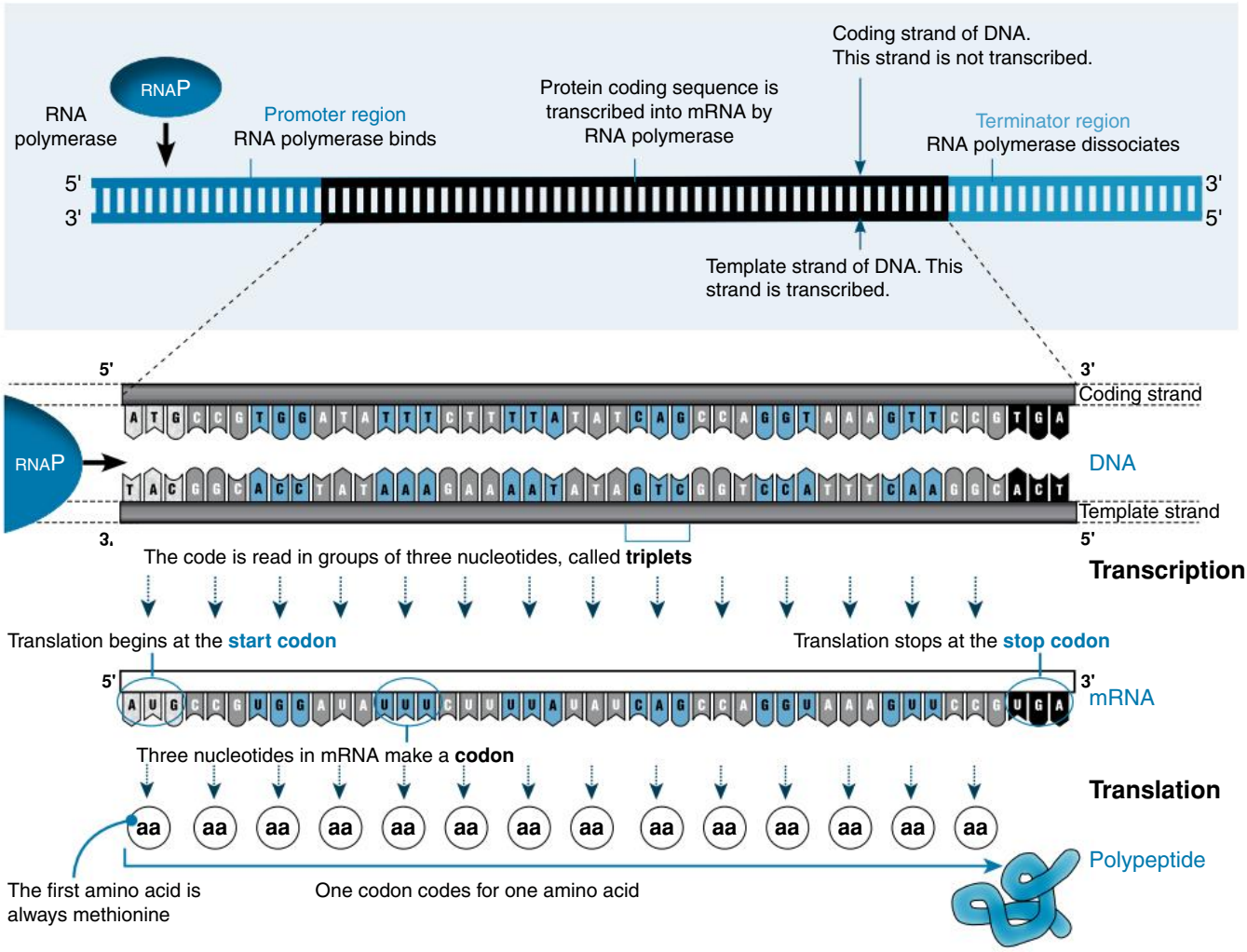
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LINK 173 LINK 172 LINK 125 KNOW

122 The Simplest Case: Genes to Proteins

Key Idea: Genes are sections of DNA that code for proteins. Genes are expressed when they are transcribed into messenger RNA (mRNA) and then translated into a protein.

Gene expression is the process of rewriting a gene into a protein. It involves **transcription** of the DNA into mRNA and **translation** of the mRNA into protein. A gene is bounded by a start (promoter) region, upstream of the gene, and a terminator region, downstream of the gene. These regions control transcription by telling RNA polymerase where to start and stop transcription of the gene. The information flow for gene to protein is shown below. Nucleotides are read in groups of three called triplets. The equivalent on the mRNA molecule is the codon. Some codons have a special control functions (start and stop) in the making of a protein.



1. (a) The three base code on DNA is called: _____

   (b) The three base code on mRNA is called: _____

2. (a) What is a **gene**? _____

   _____

   (b) What molecule transcribes the gene? _____

   (c) What is the role of the promoter and terminator regions? _____

   _____

3. What does the term **gene expression** mean? _____

   _____

4. Recall the anti-parallel nature of DNA, with the strands orientated in opposite directions. Explain its significance:

   _____

   _____

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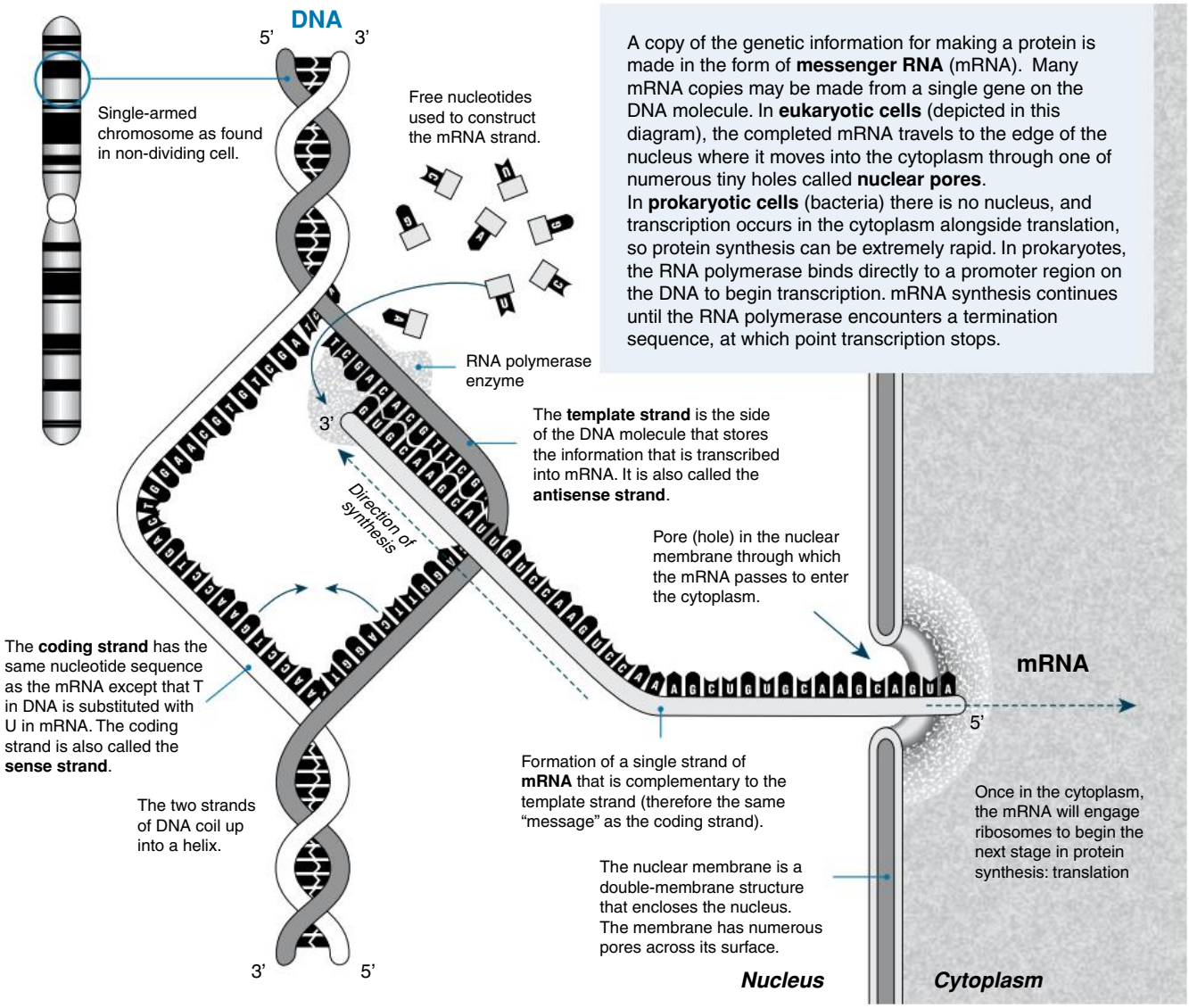
123 Transcription in Eukaryotes

Key Idea: Transcription is the first step of gene expression. A segment of DNA is transcribed (rewritten) into mRNA. In eukaryotes, transcription takes place in the nucleus.

The enzyme that directly controls transcription is RNA polymerase, which makes a strand of mRNA using the single strand of DNA (the **template strand**) as a template. The enzyme transcribes a gene length of DNA at a time and recognises start and stop signals (codes) at the beginning and end of the gene. Only RNA polymerase is involved in mRNA synthesis as it unwinds the DNA as well. It is common to find several RNA polymerase enzyme molecules on the same gene at any one time, allowing a high rate of mRNA synthesis to occur. In eukaryotes, non-coding sections called **introns** must first be removed and the remaining **exons** spliced together to form mature mRNA before the mRNA can be translated into a protein.

A copy of the genetic information for making a protein is made in the form of **messenger RNA** (mRNA). Many mRNA copies may be made from a single gene on the DNA molecule. In **eukaryotic cells** (depicted in this diagram), the completed mRNA travels to the edge of the nucleus where it moves into the cytoplasm through one of numerous tiny holes called **nuclear pores**.

In **prokaryotic cells** (bacteria) there is no nucleus, and transcription occurs in the cytoplasm alongside translation, so protein synthesis can be extremely rapid. In prokaryotes, the RNA polymerase binds directly to a promoter region on the DNA to begin transcription. mRNA synthesis continues until the RNA polymerase encounters a termination sequence, at which point transcription stops.



1. Explain the role of messenger RNA (mRNA) in protein synthesis: _____

2. The genetic code contains punctuation codons to mark the starting and finishing points of the code for synthesis of polypeptide chains and proteins. Consult the mRNA–amino acid table earlier in this workbook and state the codes for:

   (a) Start codon: _____ (b) Stop (termination) codons: _____

3. For the following triplets on the DNA, determine the **codon** sequence for the mRNA that would be synthesised:

   (a) Triplets on the DNA: T A C T A G C C G C G A T T T

   Codons on the mRNA: _____

   (b) Triplets on the DNA: T A C A A G C C T A T A A A A

   Codons on the mRNA: _____

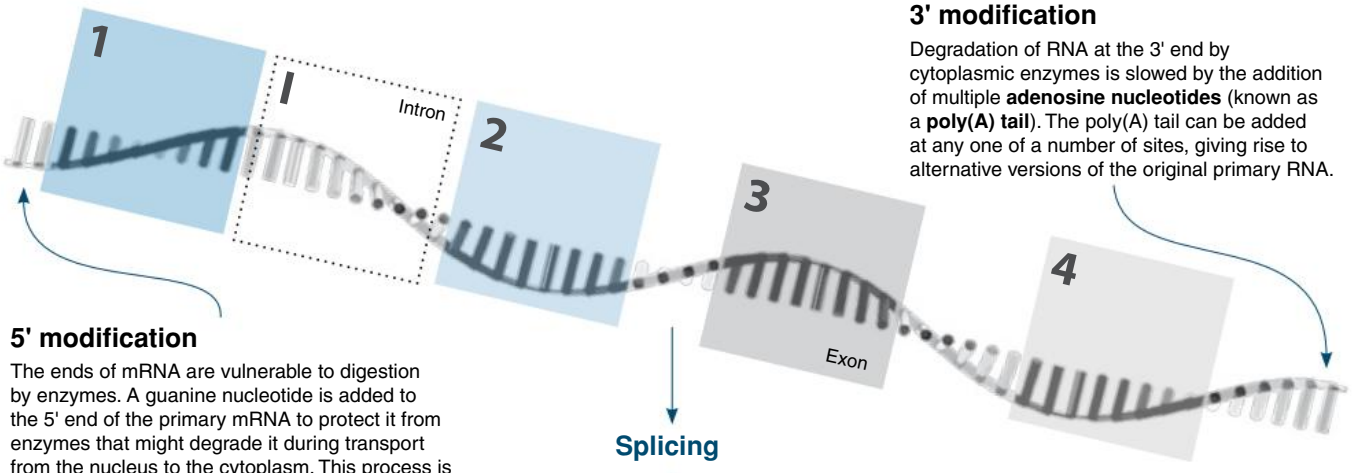
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124 Post-transcriptional Modification

Key Idea: Primary mRNA molecules are modified before they are translated into proteins.

Human DNA contains only 25,000 genes, but produces possibly up to 1 million different proteins. Each gene must therefore produce more than one protein. This is achieved by both **post transcriptional** and **post translational modification**. Primary mRNA contains exons and introns. Usually introns are removed after transcription and the exons are spliced together. However, the number of exons and the way they are spliced varies. This creates variations in the translated polypeptide chain. These mechanisms allow for the production of the diverse range of proteins.

Post Transcriptional Modification



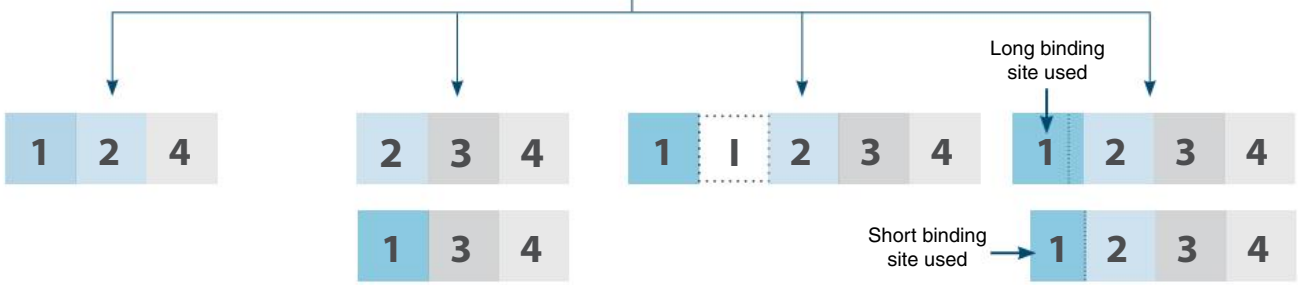
3' modification

Degradation of RNA at the 3' end by cytoplasmic enzymes is slowed by the addition of multiple **adenosine nucleotides** (known as a **poly(A) tail**). The poly(A) tail can be added at any one of a number of sites, giving rise to alternative versions of the original primary RNA.

5' modification

The ends of mRNA are vulnerable to digestion by enzymes. A guanine nucleotide is added to the 5' end of the primary mRNA to protect it from enzymes that might degrade it during transport from the nucleus to the cytoplasm. This process is called capping.

Splicing



Exon Skipping
During splicing, an exon may be skipped. This is a relatively common way to produce protein variants in mammals.

Mutually Exclusive Exons
In some cases, only one of two exons (but never both) will be incorporated into the mature mRNA.

Intron Retention
Introns are not always removed during the splicing process. In some rare cases the intron is retained in the mature mRNA.

Alternative Binding Sites
Exons may contain more than one site for binding to other exons. If the shorter version is used, the remaining code is discarded, and results in a shorter mRNA sequence.

1. Explain how so many proteins can be produced from so few genes: _____

2. Describe the ways in which mRNA can be modified to code for different proteins: _____

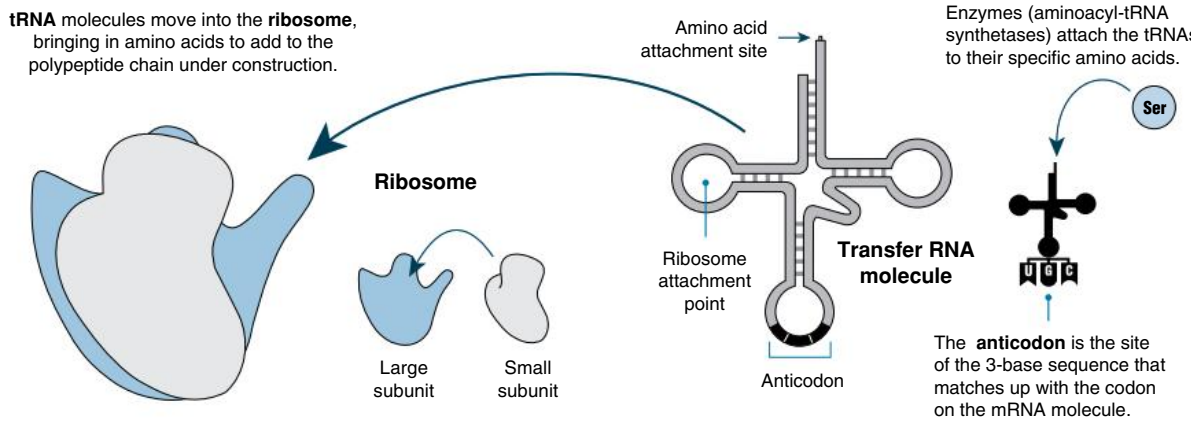
3. Explain the advantage of being able to modify the mRNA to produce different proteins: _____

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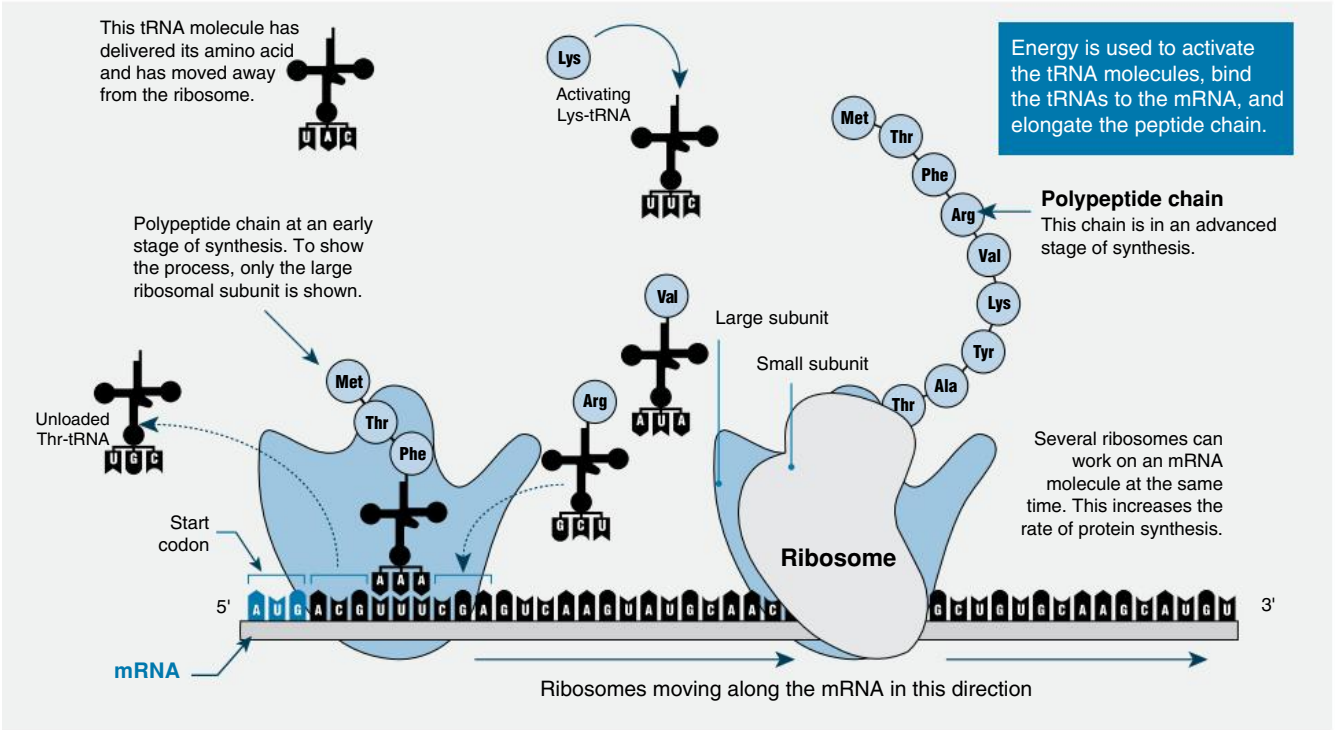
125 Translation

Key Idea: Translation is the second step of gene expression. It occurs in the cytoplasm, where ribosomes read the mRNA code and decode it to synthesise protein.

In eukaryotes, translation occurs in the cytoplasm associated with free ribosomes or ribosomes on the rough endoplasmic reticulum. The diagram below shows how a mRNA molecule can be 'serviced' by many ribosomes at the same time. The role of the tRNA molecules is to bring in the individual amino acids. The anticodon of each tRNA must make a perfect complementary match with the mRNA codon before the amino acid is released. Once released, the amino acid is added to the growing polypeptide chain by enzymes.



Ribosomes are made up of a complex of ribosomal RNA (rRNA) and proteins. They exist as two separate sub-units (above) until they are attracted to a binding site on the mRNA molecule, when they join together. Ribosomes have binding sites that attract transfer RNA (**tRNA**) molecules loaded with amino acids. The tRNA molecules are about 80 nucleotides in length and are made under the direction of genes in the chromosomes. There is a different tRNA molecule for each of the different possible anticodons (see the diagram below) and, because of the degeneracy of the genetic code, there may be up to six different tRNAs carrying the same amino acid.



1. For the following codons on the mRNA, determine the **anticodons** for each tRNA that would deliver the amino acids:

   Codons on the mRNA: U A C U A G C C G C G A U U U

   Anticodons on the tRNAs: _____

2. There are many different types of tRNA molecules, each with a different anticodon (HINT: see the mRNA table).

   (a) How many different tRNA types are there, each with a unique anticodon? _____

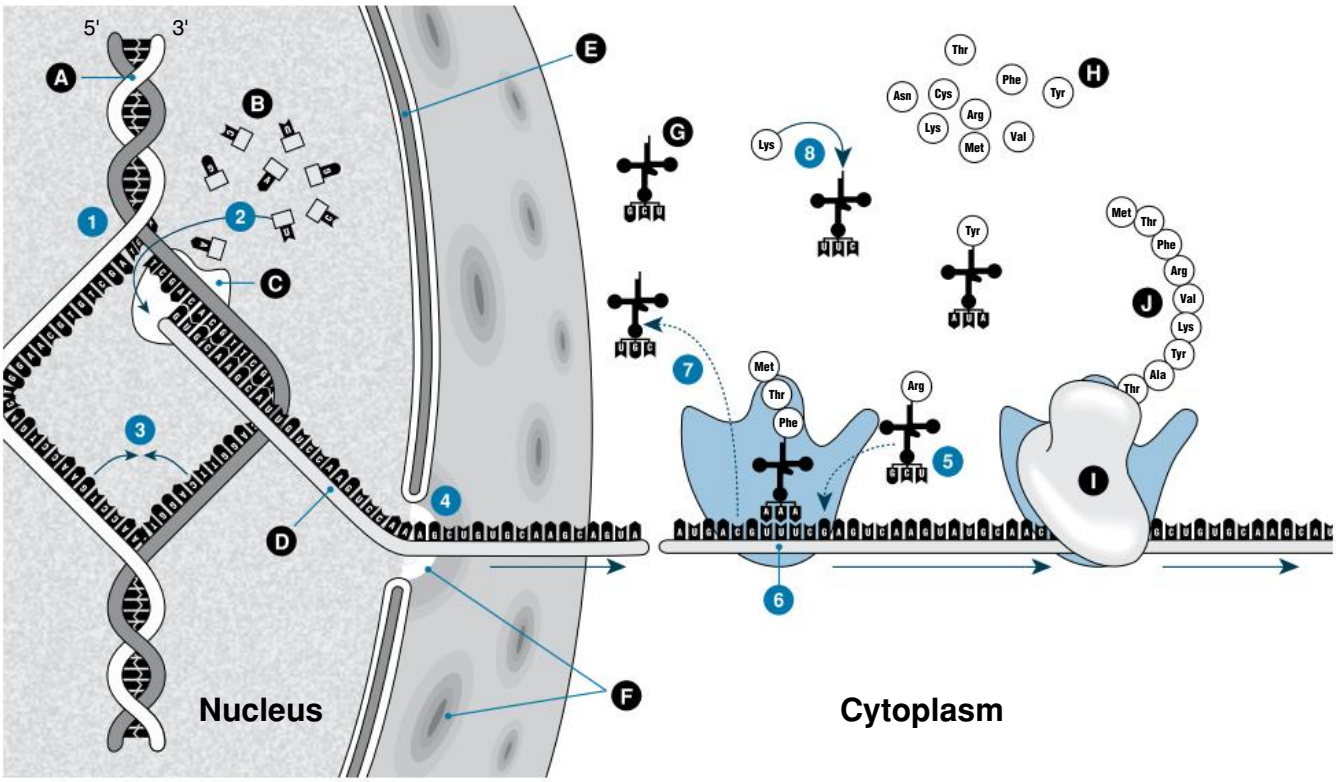
   (b) Explain your answer: _____

   _____

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LINK 122 LINK 119 WEB 125 KNOW

126 Protein Synthesis Summary



The diagram above shows an overview of the process of protein synthesis. It is a combination of the diagrams from the previous two pages. Each of the major steps in the process are numbered, while structures are labelled with letters.

1. Briefly describe each of the numbered processes in the diagram above:

   (a) Process 1: _____

   (b) Process 2: _____

   (c) Process 3: _____

   (d) Process 4: _____

   (e) Process 5: _____

   (f) Process 6: _____

   (g) Process 7: _____

   (h) Process 8: _____

2. Identify each of the structures marked with a letter and write their names below in the spaces provided:

   (a) Structure A: _____ (f) Structure F: _____

   (b) Structure B: _____ (g) Structure G: _____

   (c) Structure C: _____ (h) Structure H: _____

   (d) Structure D: _____ (i) Structure I: _____

   (e) Structure E: _____ (j) Structure J: _____

3. Describe two factors that would determine whether or not a particular protein is produced in the cell:

   (a) _____

   _____

   (b) _____

   _____

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127 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its correct definition, as identified by its preceding letter code.

allele

anticodon

autosome

base-pairing rule

chromatid

chromosome

coding strand

diploid

DNA

exons

gene expression

genetic code

hydrogen bonding

introns

karyotype

nucleic acids

nucleotides

protein

purine

pyrimidine

RNA (mRNA, rRNA, tRNA)

sex chromosome

template strand

transcription

translation

A Single stranded nucleic acid that consists of nucleotides that contain ribose sugar.

B Organic macromolecules composed of linear chains of amino acids joined together by peptide bonds and then organised, e.g. through folding, into a functional structure.

C The process by which genetic information is used to produce a functional gene product.

D A set of rules by which information encoded in DNA or mRNA is translated into proteins.

E The rule governing the pairing of complementary bases in DNA.

F The region of a transfer RNA with a sequence of three bases that are complementary to a codon in the messenger RNA.

G DNA regions within a gene that are not translated into protein.

H A two-ringed organic base that forms adenine and guanine in nucleic acids.

I The number and appearance of chromosomes in the nucleus of a eukaryotic cell.

J The process of creating an equivalent RNA copy of a sequence of DNA.

K Single piece of DNA that contains many genes and associated regulatory elements and proteins. Found within the nucleus in eukaryotes and as a singular circular piece in prokaryotes.

L Form of intermolecular bonding between hydrogen and an electronegative atom such as oxygen.

M The sequence of DNA that is read during the synthesis of mRNA.

N Universally found macromolecules composed of chains of nucleotides. These molecules carry genetic information within cells.

O The DNA strand with the same base sequence as the RNA transcript produced (although with thymine replaced by uracil in mRNA).

P A non-sex chromosome.

Q Macromolecule consisting of many millions of units containing a phosphate group, sugar and a base (A,T, C or G). Stores the genetic information of the cell.

R The structural units of nucleic acids, DNA and RNA.

S A single-ringed organic base that forms uracil, cytosine, or thymine in nucleic acids.

T Having two homologous copies of each chromosome (2N), usually one from the mother and one from the father.

U Nucleic acid sequences that are represented in the mature form of an RNA molecule.

V The chromosome that carries the gene for determination of sex in individual organisms.

W The stage of gene expression in which mRNA is decoded to produce a polypeptide.

X One of a pair of duplicated chromosomes produced prior to cell division, joined at the centromere.

Y One of a number of alternative versions of a gene that can occupy a given genetic locus on a chromosome.

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QLD As required
VCE Unit 4 (1)
WA ATAR Unit 3
SA Theme C

Cell Division

Key terms

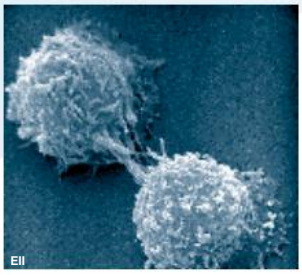
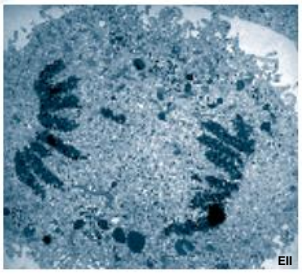
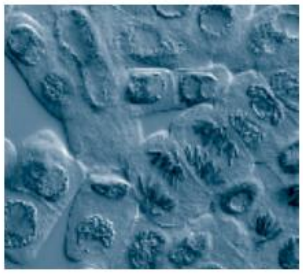
adult stem cell (ASC)
anaphase
asexual reproduction
binary fission
cell cycle
cellular differentiation
crossing over
cytokinesis
DNA ligase
DNA polymerase
DNA replication
embryonic stem cell (ESC)
first gap phase (G1)
gene regulation
helicase
meiosis
metaphase
mitosis
Okazaki fragments
pluripotent
potency
prophase
second gap phase (G2)
self-renewal
stem cells
telophase
totipotent

DNA replication

Learning aims and skills

Activity number

- ☐ 1 Explain the semi-conservative replication of DNA during interphase, including the role of DNA polymerase, and DNA helicase. — 128 129
- ☐ 2 Explain the significance of DNA replication in the 5' to 3' direction. Relate this to the formation of the leading strand and the lagging strand. — 129
- ☐ 3 Analyse the results of Meselson and Stahl's experiments on DNA replication. — 130 131
- ☐ 4 Recognise DNA replication as a necessary precursor to cell division. — 133

Cell reproduction

Learning aims and skills

Activity number

- ☐ 5 Describe asexual reproduction by binary fission in prokaryotes. — 132
- ☐ 6 Describe the cell cycle in eukaryotes, including reference to: DNA replication (S), growth (G1 and G2), and mitosis (M phase). — 133
- ☐ 7 Describe stages in mitosis: prophase, metaphase, anaphase, and telophase. Recognise these stages in light and electron micrographs.. — 133
- ☐ 8 Describe and explain cytokinesis in both plant cells and animal cells. — 133
- ☐ 9 Describe how the cell cycle is regulated according to requirements. Recall the problems that occur when errors in regulation occur. — 134
- ☐ 10 Distinguish between meiosis and mitosis in terms of their cellular outcomes. — 139

Stem cells

Learning aims and skills

Activity number

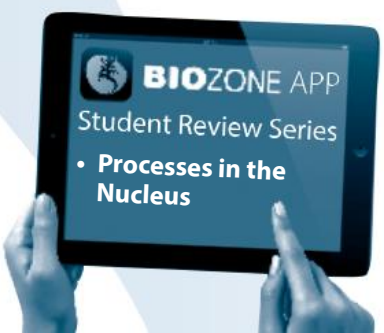
- ☐ 11 Describe the properties of stem cells, including self-renewal and potency. Describe the usual role of stem cells in multicellular organisms. — 135
- ☐ 12 Distinguish between totipotent and pluripotent cells. Distinguish between embryonic stem cells and adult stem cells with respect to potency. — 135

Meiosis and variation

Learning aims and skills

Activity number

- ☐ 13 Describe the events in the two divisions of meiosis. Explain the role of crossing over and recombination in creating new combinations of alleles in the gametes. — 136-138
- ☐ 14 Explain why a reduction division is necessary prior to fertilisation in sexually reproducing organisms. — 136



128 DNA Replication

Key Idea: Semi conservative DNA replication produces two identical copies of DNA, each containing half original material and half new material.

Before a cell can divide, it must double its DNA. It does this by a process called DNA replication. This process ensures that each resulting cell receives a complete set of genes from the original cell. After the DNA has replicated, each chromosome is made up of two chromatids, joined at the centromere. The two chromatids will become separated during cell division to form two separate chromosomes. During DNA replication, nucleotides are added at the replication fork. Enzymes are responsible for all of the key events.

Step 1
Unwinding the DNA molecule

A normal chromosome consists of an unreplicated DNA molecule. Before cell division, this long molecule of double stranded DNA must be replicated.

For this to happen, it is first untwisted and separated (unzipped) at high speed at its replication fork by an enzyme called helicase. Another enzyme relieves the strain that this generates by cutting, winding and rejoining the DNA strands.

Step 2
Making new DNA strands

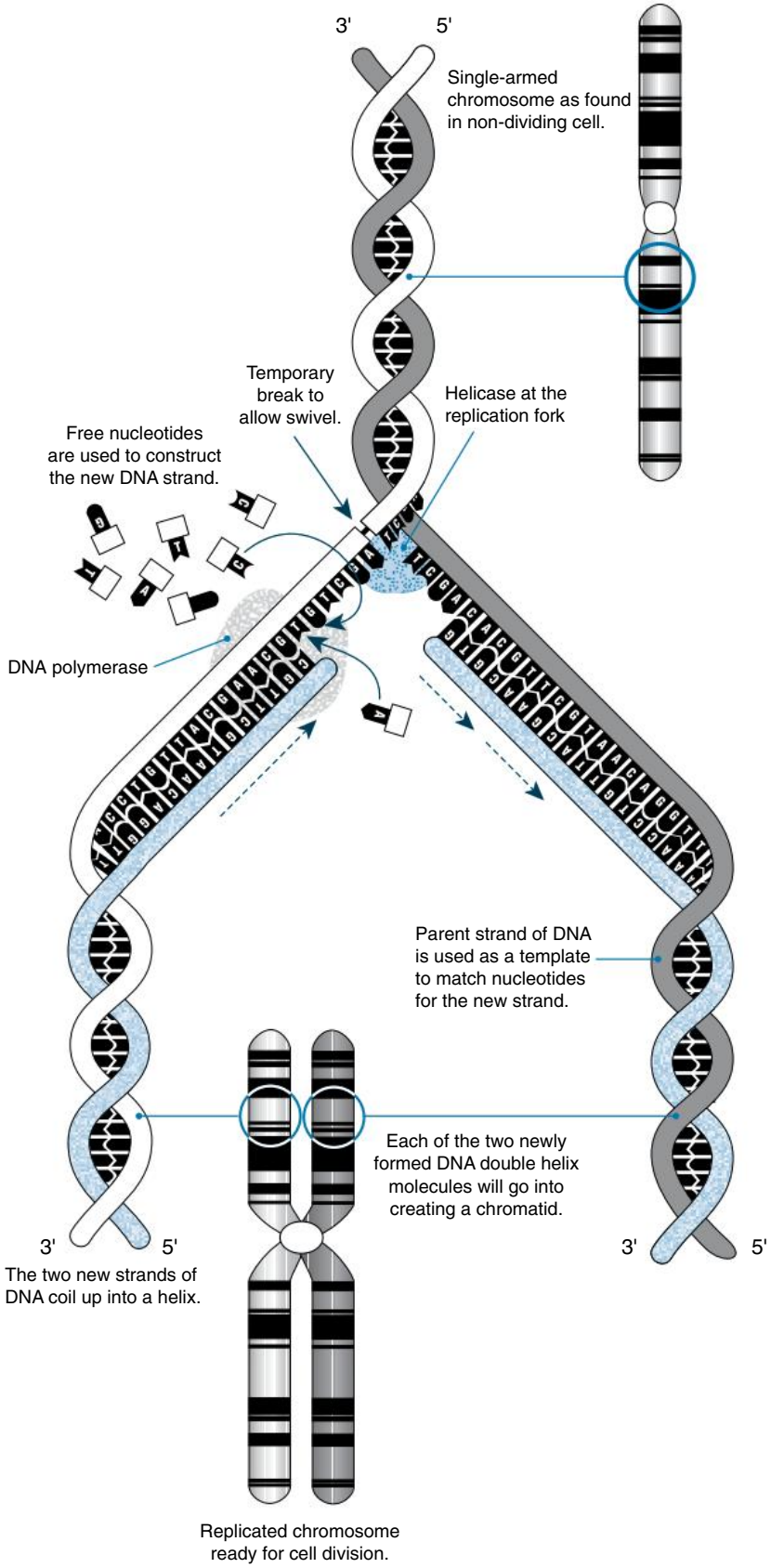
The formation of new DNA is carried out mostly by an enzyme complex called **DNA polymerase**.

DNA polymerase works in a 5' to 3' direction so nucleotides are assembled in a continuous fashion on one strand but in short fragments on the other strand. These fragments are later joined by an enzyme to form one continuous length.

Step 3
Rewinding the DNA molecule

Each of the two new double-helix DNA molecules has one strand of the original DNA (dark gray and white) and one strand that is newly synthesised (blue). The two DNA molecules rewind into their double-helix shape again.

DNA replication is semi-conservative, with each new double helix containing one old (parent) strand and one newly synthesised (daughter) strand. The new chromosome has twice as much DNA as a non-replicated chromosome. The two chromatids will become separated in the cell division process to form two separate chromosomes.



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1. What is the purpose of DNA replication? _____

2. Summarise the three main steps involved in DNA replication:

   (a) _____

   (b) _____

   (c) _____

3. For a cell with 22 chromosomes, state how many chromatids would exist following DNA replication: _____

4. What does it mean when we say DNA replication is semi-conservative? _____

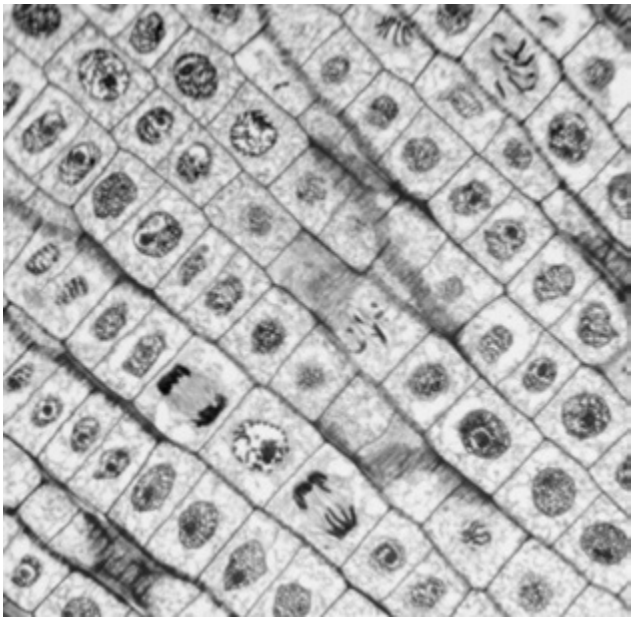
5. DNA replication occurs during the S (synthesis) phase of the **cell cycle**. This is part of a larger phase called interphase. It is the phase in which the cell is not dividing (in mitosis).

   The light micrograph (right) shows a section of cells in an onion root tip. These cells have a cell cycle of approximately 24 hours. The cells can be seen to be in various stages of the cell cycle. By counting the number of cells in the various stages it is possible to calculate how long the cell spends in each stage of the cycle.

   Count and record the number of cells in the image which are undergoing mitosis and those that are in interphase. Estimate the amount of time a cell spends in each phase.

Stage	No. of cells	% of total cells	Estimated time in stage
Interphase			
Mitosis			
Total		100	

Onion Root Tip Cells



6. Match the statements in the table below to form complete sentences, then put the sentences in order to make a coherent paragraph about DNA replication and its role:

The enzymes also proofread the DNA during replication...	...is required before mitosis or meiosis can occur.
DNA replication is the process by which the DNA molecule...	...by enzymes.
Replication is tightly controlled...	...to correct any mistakes.
After replication, the chromosome...	...and half new DNA.
DNA replication...	...during mitosis.
The chromatids separate...	...is copied to produce two identical DNA strands.
A chromatid contains half original ...	...is made up of two chromatids.

Write the complete paragraph here: _____

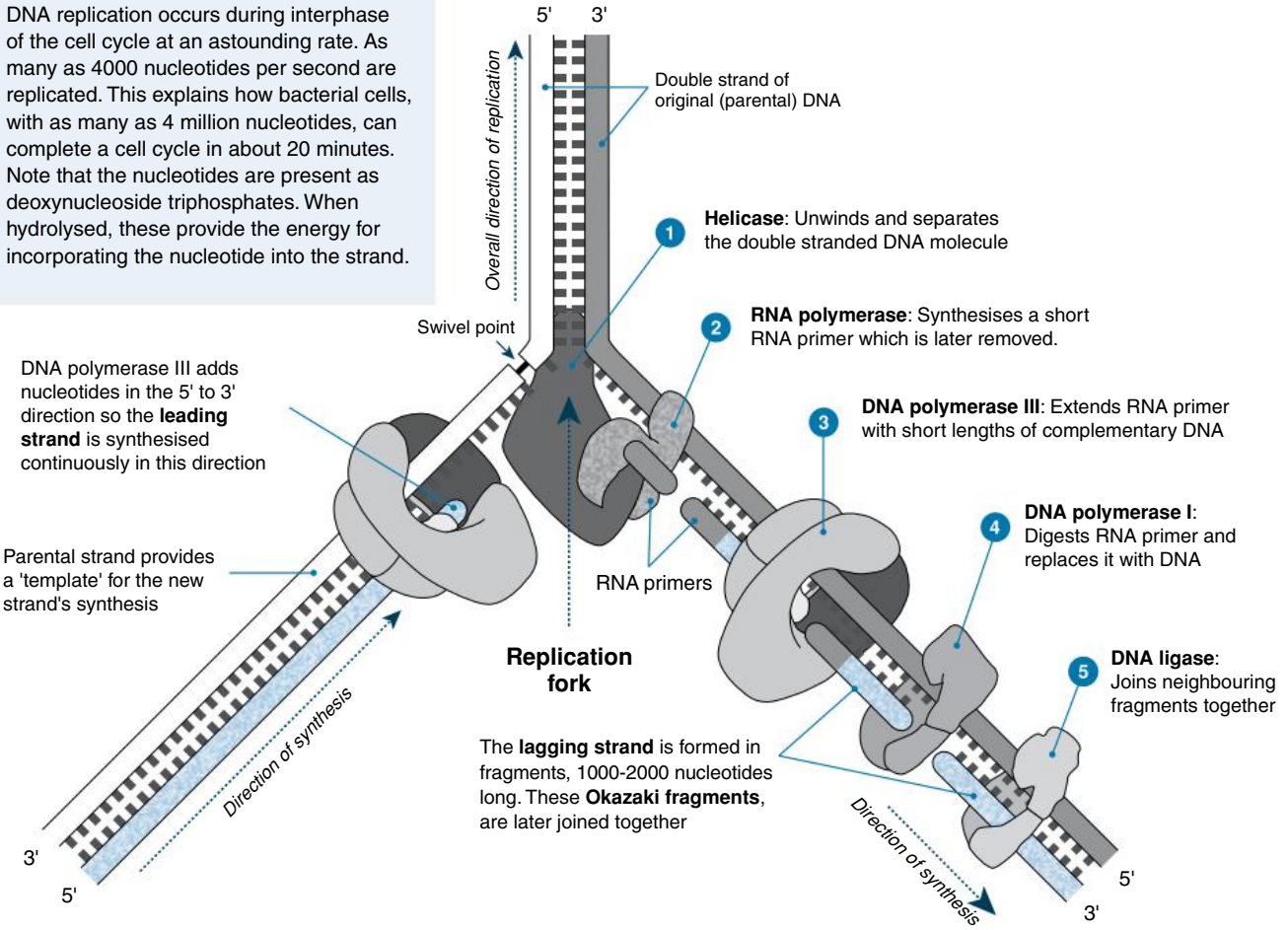
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129 Enzyme Control of DNA Replication

Key Idea: DNA replication is a directional process controlled by several different enzymes.

DNA replication involves many enzyme-controlled steps. They are shown below as separate, but many of the enzymes are clustered together as enzyme complexes. As the DNA is replicated, enzymes 'proof-read' it and correct mistakes. The polymerase enzyme can only work in one direction, so that one new strand is constructed as a continuous length (the leading strand) while the other new strand (the lagging strand) is made in short segments to be later joined together.

DNA replication occurs during interphase of the cell cycle at an astounding rate. As many as 4000 nucleotides per second are replicated. This explains how bacterial cells, with as many as 4 million nucleotides, can complete a cell cycle in about 20 minutes. Note that the nucleotides are present as deoxynucleoside triphosphates. When hydrolysed, these provide the energy for incorporating the nucleotide into the strand.



1. Describe the general role of enzymes in DNA replication: _____

2. State the specific role of each of the following enzymes in DNA replication:

   (a) Helicase: _____

   (b) DNA polymerase I: _____

   (c) DNA polymerase III: _____

   (d) Ligase: _____

3. Determine the time it would take for a bacterium to replicate its DNA (see note in diagram above): _____

4. How is the energy for incorporating the nucleotides into the strand provided? _____

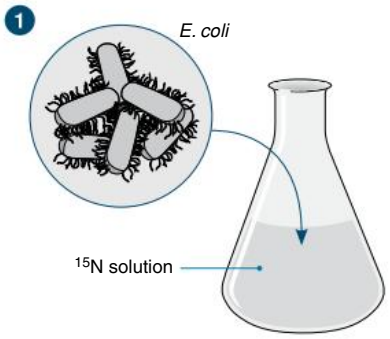
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130 Meselson and Stahl's Experiment

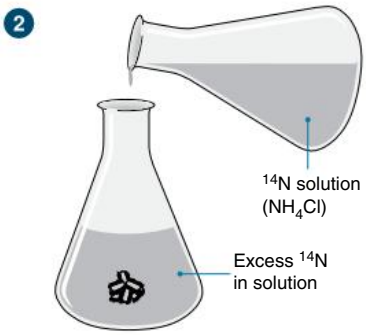
Key Idea: Meselson and Stahl devised an experiment that showed DNA replication is semi-conservative. The anti-parallel, complementary structure of DNA suggested three possible mechanisms for its replication. The **semi-conservative model** proposed that each strand served as a template, forming new DNA molecules that were half old and half new DNA. The **conservative model** proposed that the original DNA served as a complete template so that the new DNA comprised two new strands. The **dispersive model** proposed that the two new DNA molecules had new and old DNA mixed throughout them. **Meselson and Stahl** devised a simple experiment to determine which model was correct.

Meselson and Stahl's Experiment

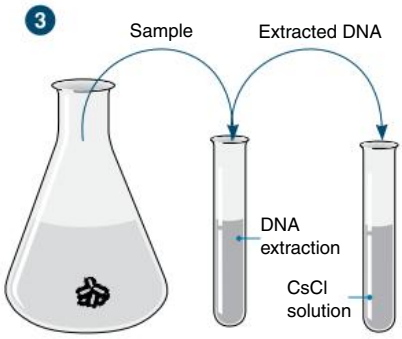
E. coli were grown for several generations in a medium containing a **heavy nitrogen isotope** (^{15}N). Once all the bacterial DNA contained ^{15}N, they were transferred to a medium containing a **light nitrogen isotope** (^{14}N). After the transfer, newly synthesised DNA would contain ^{14}N and old DNA would contain ^{15}N.



E. coli were grown in a nutrient solution containing ^{15}N. After 14 generations, all the bacterial DNA contained ^{15}N. A sample is removed. This is **generation 0**.



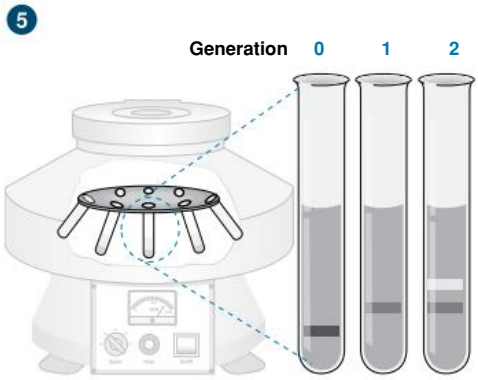
Generation 0 is added to a solution containing excess ^{14}N (as NH_4Cl). During replication, new DNA will incorporate ^{14}N and be 'lighter' than the original DNA (which contains only ^{15}N).



Every generation (~ 20 minutes), a sample is taken and treated to release the DNA. The DNA is placed in a CsCl solution which provides a density gradient for separation of the DNA.



Samples are spun in a high speed ultracentrifuge at 140,000 *g* for 20 hours. Heavier ^{15}N DNA moves closer to the bottom of the test tube than light ^{14}N DNA or intermediate ^{14}N/ ^{15}N DNA.



All the DNA in the generation 0 sample moved to the bottom of the test tube. All the DNA in the generation 1 sample moved to an intermediate position. At generation 2 half the DNA was at the intermediate position and half was near the top of the test tube. In subsequent generations, more DNA was near the top and less was in the intermediate position.

Models for DNA Replication



Conservative | Semi-conservative | Dispersive

1. Describe each of the DNA replication models:

   (a) Conservative: _____

   _____

   (b) Semi-conservative: _____

   _____

   (c) Dispersive: _____

   _____

2. Explain why the *E. coli* were grown in an ^{15}N solution before being transferred to an ^{14}N solution: _____

   _____

   _____

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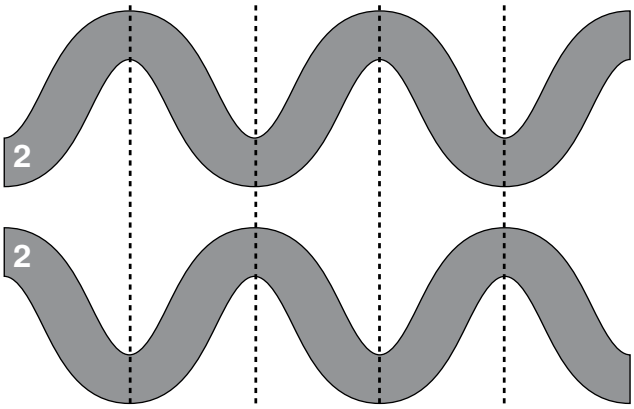
131 Modelling DNA Replication

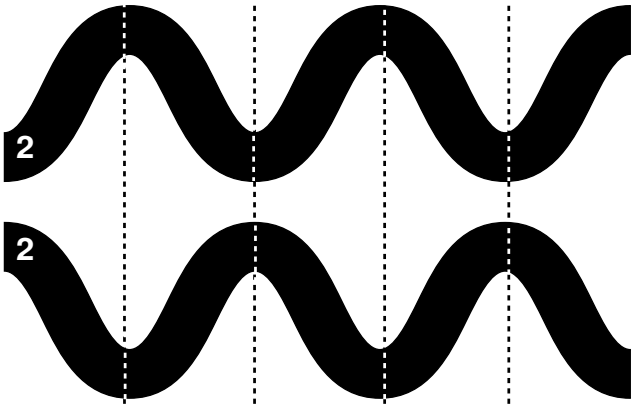
Key Idea: Meselson and Stahl's experiment to determine the nature of DNA replication can be modelled.

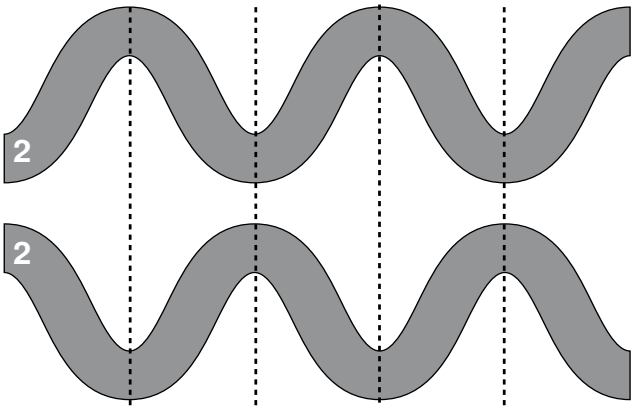
There were three possible models proposed to explain how DNA replicated. Meselson and Stahl's experiment was able to determine which method was used by starting with parent DNA that was heavier than would normally be expected. They were then able to analyse the relative weight of the replicated DNA to work out the correct replication method.

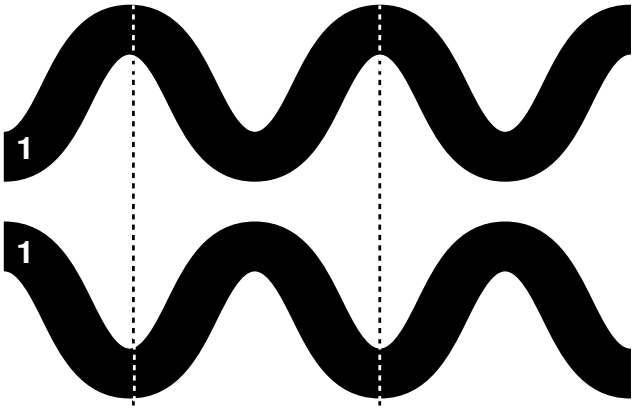
Instructions:

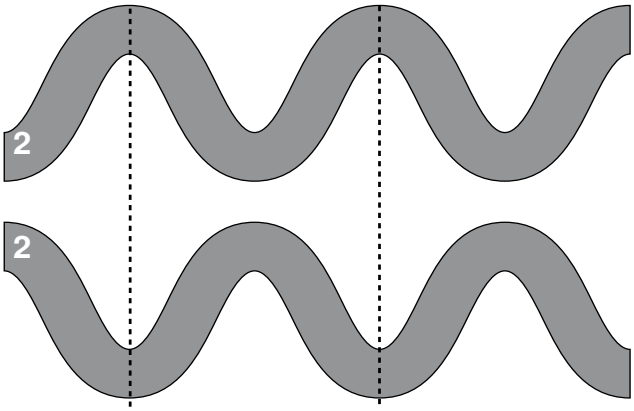
1. Cut out the DNA shapes provided on this page.
2. Intertwine the first pair (labelled 0) of heavy ^{15}N (black) DNA. This forms Generation 0 (parental DNA).
3. Use the descriptions of the three possible models for DNA replication on the previous page to model DNA replication in semi-conservative, conservative, and dispersive DNA replication.
4. For each replication method, record in the spaces provided on page 175 the percentage of **heavy** ^{15}N-^{15}N (black-black), **intermediate** ^{15}N-^{14}N (black-grey), **light** ^{14}N-^{14}N (grey-grey), or other DNA molecules formed.
5. For the dispersive model you will need to cut the DNA along the dotted lines and then stick them back together in the dispersed sequence with tape. **Construct the dispersive model LAST.**

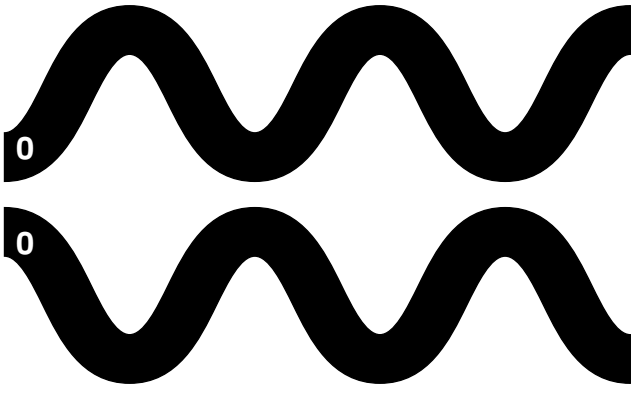


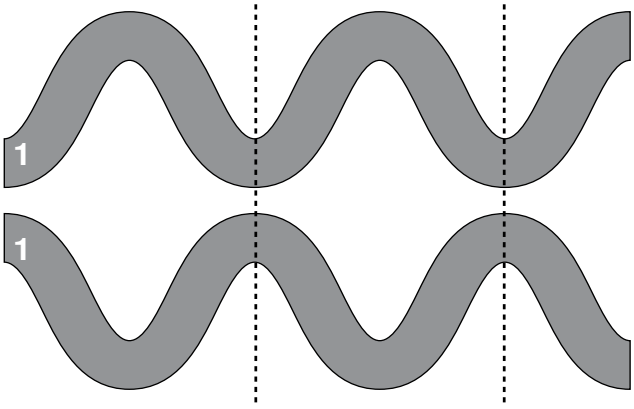












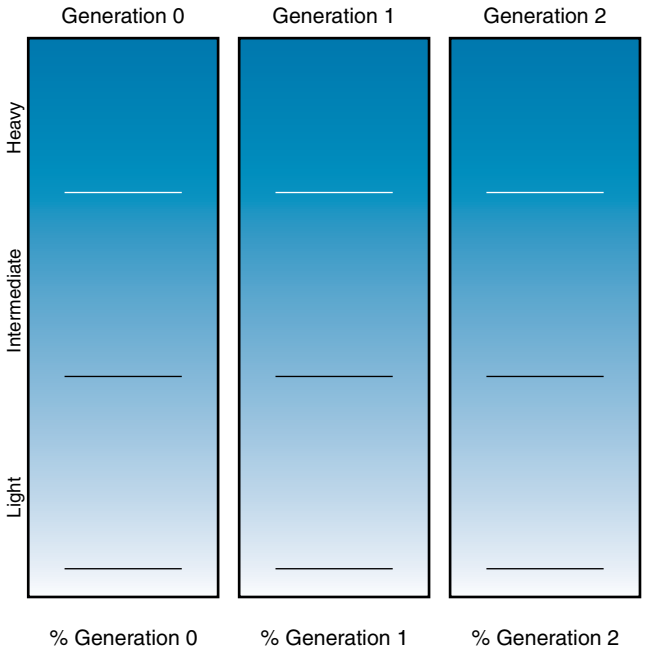
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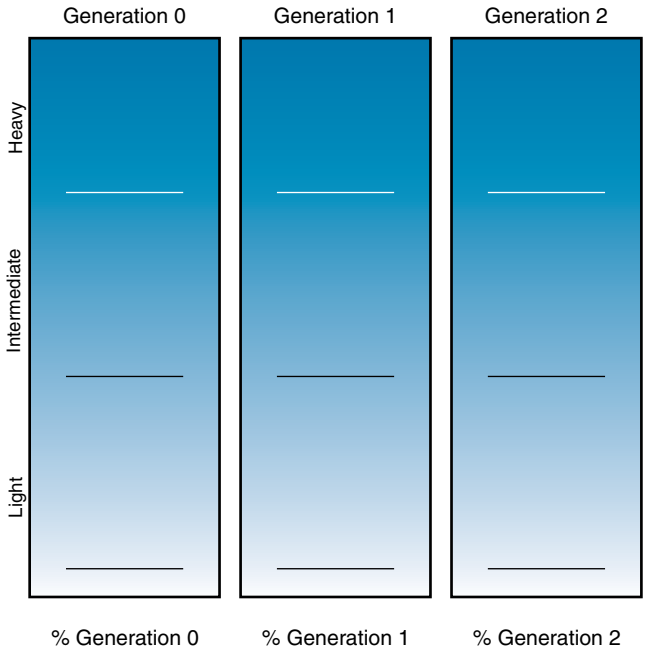


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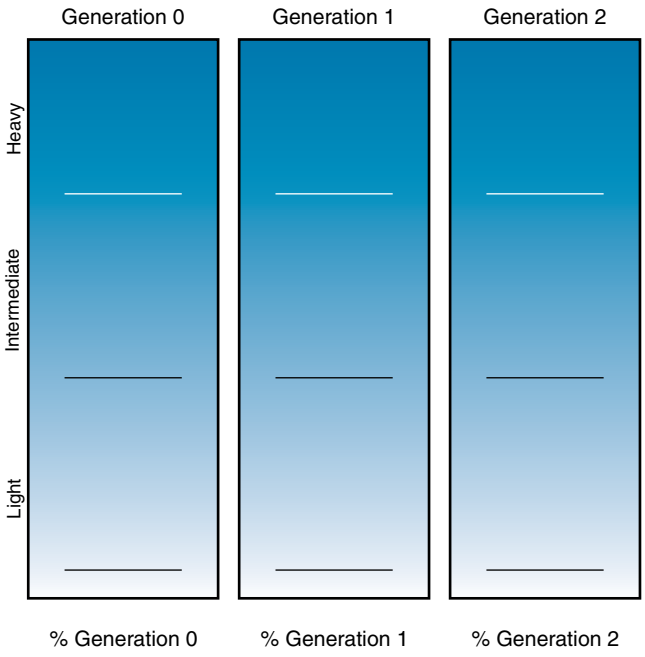
Conservative

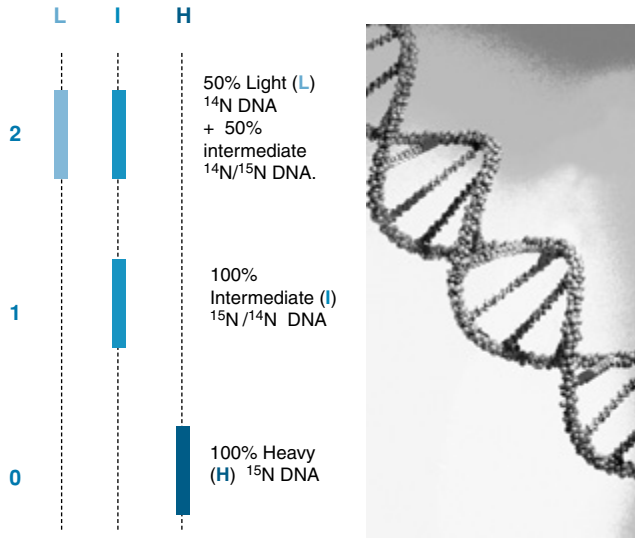


Semi-conservative



Dispersive





The results from Meselson's and Stahl's are shown graphically above. All of the generation 1 DNA contained one light strand (^{14}N) and one heavy strand (^{15}N) to produce an intermediate density. At generation 2, 50% of the DNA was light and 50% was intermediate DNA.

1. (a) Compare your modelling results to the results gained by Meselson and Stahl to decide which of the three DNA replication models is supported by the data:

_____

_____

(b) Was Watson and Crick's proposal correct? _____

2. Identify the replication model that fits the following data:

(a) 100% of generation 0 is "heavy DNA", 50% of generation 1 is "heavy" and 50% is "light", and 25% of generation 2 is "heavy" and 75% is "light":

_____

(b) 100% of generation 0 is "heavy DNA", 100% of generation 1 is "intermediate DNA", and 100% generation 2 lies between the "intermediate" and "light" DNA regions:

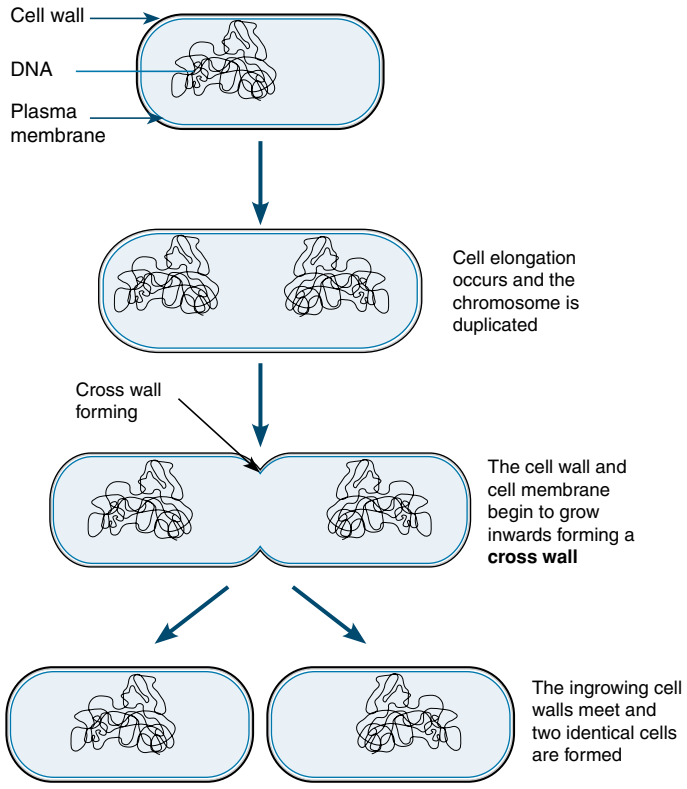
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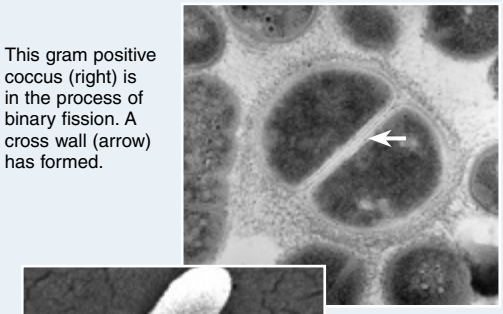
132 Binary Fission

Key Idea: Binary fission involves division of the parent body into two, fairly equal, parts to produce two identical cells.

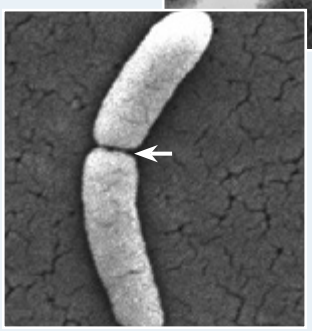
Binary fission is a form of asexual reproduction carried out by most prokaryotes, some eukaryotic organelles, such as chloroplasts, and some unicellular eukaryotes (although the process is somewhat different in eukaryotic cells). The time required for a bacterial cell to divide, or for a population of bacterial cells to double, is called the generation time. Generation times may be quite short (20 minutes) in some species and as long as several days in others.



Most bacteria reproduce asexually by binary fission (left). The cell's DNA is replicated and each copy attaches to a different part of the plasma membrane. When the cell begins to pull apart, the replicate and original chromosomes are separated. Binary fission in bacteria does not involve mitosis or cytokinesis.



This gram positive coccus (right) is in the process of binary fission. A cross wall (arrow) has formed.



This *Salmonella typhimurium* bacterium (left) has completed cell division. The separation between the two cells can be clearly seen (arrow).

CDC

Generation time (minutes)	Population size
0	1
20	2
40	4
60	8
80	
100	
120	
140	
160	
180	
200	
220	
240	
260	
280	
300	
320	
340	
360	

1. What is **binary fission**? _____

2. Explain why the formation of the **cross wall** is important in binary fission:

3. Explain the term **generation time**: _____

4. A species of bacteria reproduces every 20 minutes. Complete the table (left) by calculating the number of bacteria present at 20 minute intervals.

5. State how many bacteria were present after:

   (a) 1 hour: _____

   (b) 3 hours: _____

   (c) 6 hours: _____

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133 Mitosis and the Cell Cycle

Key Idea: Mitosis is an important part of the cell cycle in which the replicated chromosomes are separated and the cell divides, producing two new identical cells.

Mitosis (or M-phase) is part of the **cell cycle** in which an existing cell (the parent cell) divides into two (the daughter cells). Unlike meiosis, mitosis does not result in a change of chromosome numbers and the daughter cells are identical to the parent cell. Although mitosis is part of a continuous cell cycle, it is often divided into stages to help differentiate the processes occurring. Mitosis is one of the shortest stages of the cell cycle. When a cell is not undergoing mitosis, it is said to be in interphase. Interphase accounts for 90% of the cell cycle. Cytokinesis (the division of the newly formed cells) is distinct from nuclear division.

The Cell Cycle

Interphase

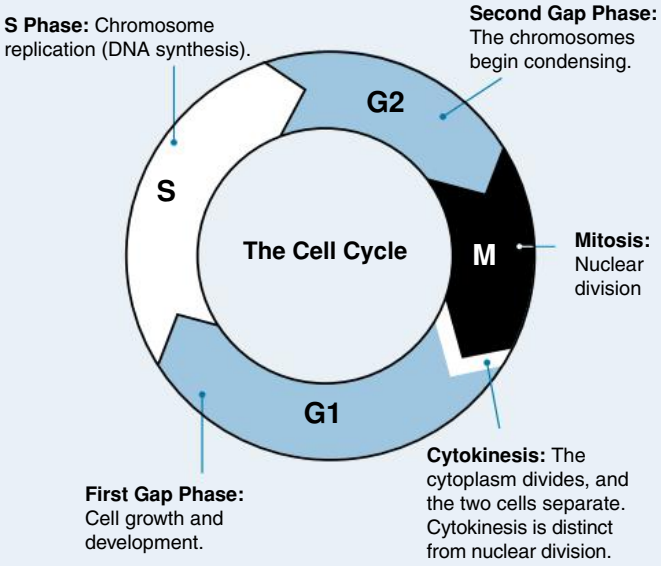
Cells spend most of their time in interphase. Interphase is divided into three stages (right):

- The first gap phase.
- The S-phase.
- The second gap phase.

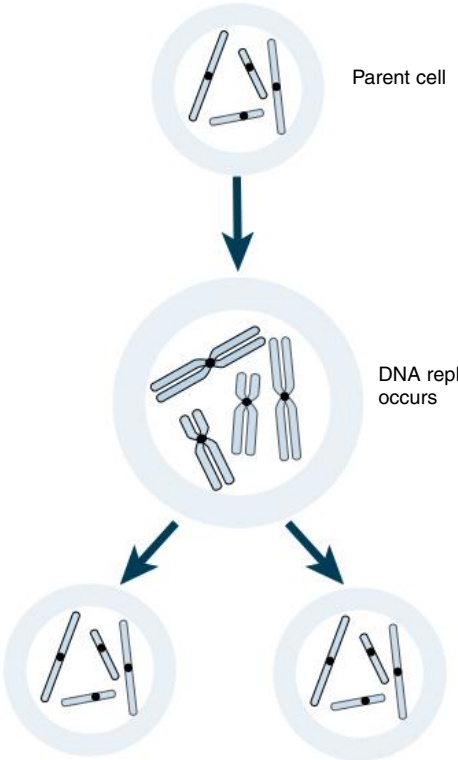
During interphase the cell grows, carries out its normal activities, and replicates its DNA in preparation for cell division.
Interphase is not a stage in mitosis.

Mitosis and cytokinesis (M-phase)

Mitosis and cytokinesis occur during M-phase. During mitosis, the cell nucleus (containing the replicated DNA) divides in two equal parts. Cytokinesis occurs at the end of M-phase. During cytokinesis the cell cytoplasm divides, and two new daughter cells are produced.



An Overview of Mitosis

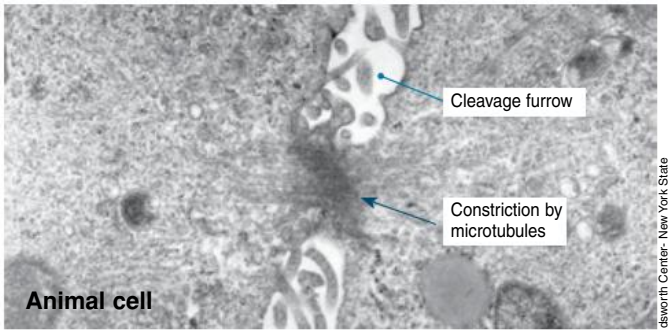


The cell divides forming two identical daughter cells. The chromosome number remains the same as the parent cell.

Cytokinesis

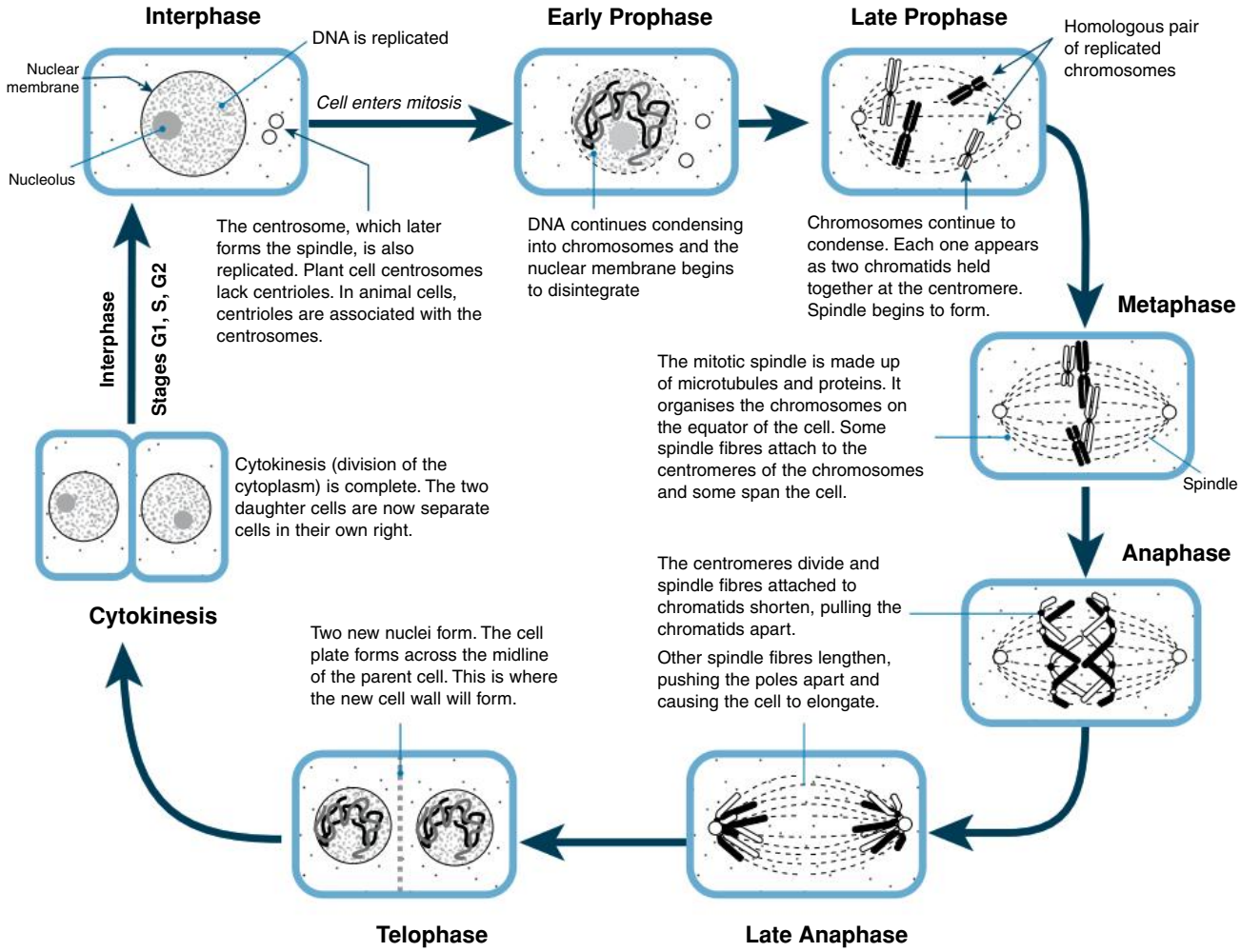
In plant cells (below top), cytokinesis (division of the cytoplasm) involves construction of a cell plate (a precursor the new cell wall) in the middle of the cell. The cell wall materials are delivered by vesicles derived from the Golgi. The vesicles coalesce to become the plasma membranes of the new cell surfaces. Animal cell cytokinesis (below bottom) begins shortly after the sister chromatids have separated in anaphase of mitosis. A contractile ring of microtubular elements assembles in the middle of the cell, next to the plasma membrane, constricting it to form a cleavage furrow. In an energy-using process, the cleavage furrow moves inwards, forming a region of abscission (separation) where the two cells will separate.





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The Cell Cycle and Stages of Mitosis



1. What is the purpose of mitosis? _____

2. What must occur before mitosis takes place? _____

3. Briefly outline what occurs during the following phases of the cell cycle:

   (a) Interphase: _____

   (b) Mitosis: _____

   (c) Cytokinesis: _____

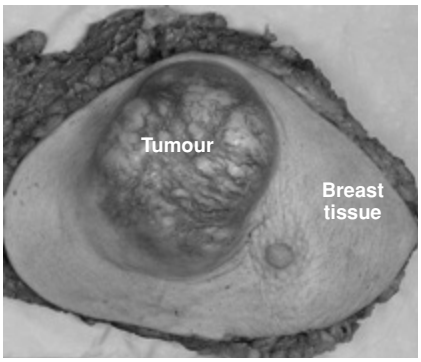
4. How is cytokinesis different in animals compared to plants? _____

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134 Regulation of the Cell Cycle

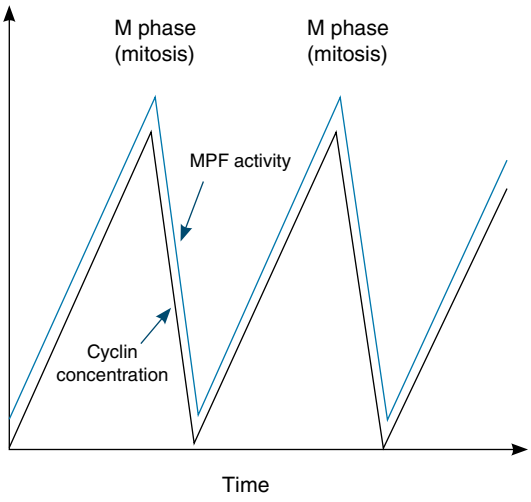
Key Idea: The cell cycle is regulation by the molecules cyclin and cyclin-dependent kinase.

Mitosis is virtually the same for all eukaryotes but aspects of the cell cycle can vary enormously between species and even between cells of the same organism. For example, the length of the cell cycle varies between cells such as intestinal and liver cells. Intestinal cells divide around twice a day, while cells in the liver divide once a year. However, if these tissues are damaged, cell division increases rapidly until the damage is repaired. Variation in the length of the cell cycle can be explained by regulatory mechanisms that slow down or speed up the cell cycle in response to changing conditions.



Emmanuelm cc3.0

Regulation of the cell cycle is important in detecting and repairing of genetic damage, and preventing uncontrolled cell division. Tumours and cancers, such as this breast cancer (above) are the result of uncontrolled cell division.



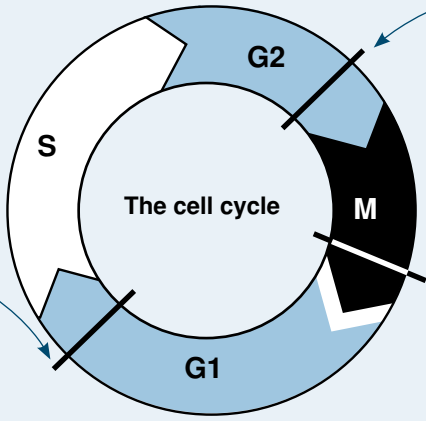
A substance called an M-phase promoting factor (MPF) controls cell regulation. MPF is made up of two regulatory molecules, **cyclins** and **cyclin-dependent kinases** (CDKs).

Cyclins are proteins that control the progression of cells through the cell cycle by activating CDKs (which are enzymes).

CDKs phosphorylate other proteins to signal a cell is ready to proceed to the next stage in the cell cycle. Without cyclin, CDK has little kinase activity; only the cyclin-CDK complex is active. CDK is constantly present in the cell, cyclin is not.

Checkpoints During the Cell cycle

There are three **checkpoints** during the cell cycle. A checkpoint is a critical regulatory point in the cell cycle. At each checkpoint, a set of conditions determines whether or not the cell will continue into the next phase. For example, cell size is important in regulating whether or not the cell can pass through the G_1 checkpoint.



G_2 Checkpoint:
Pass this checkpoint if:
- Cell size is large enough.
- Replication of chromosomes has been successfully completed.

G_1 checkpoint
Pass this checkpoint if:
- Cell size is large enough.
- Sufficient nutrients are available.
- Signals from other cells have been received.

Metaphase checkpoint
Pass this checkpoint if:
- All chromosomes are attached to the mitotic spindle.

1. What would happen if the cell cycle was not regulated? _____

_____

_____

2. (a) Suggest why the cell cycle is shorter in epithelial cells (such as intestinal cells) than in liver cells:

_____

_____

_____

(b) Describe another situation in which the cell cycle shortens to allow for a temporary rapid rate of cell division:

_____

_____

_____

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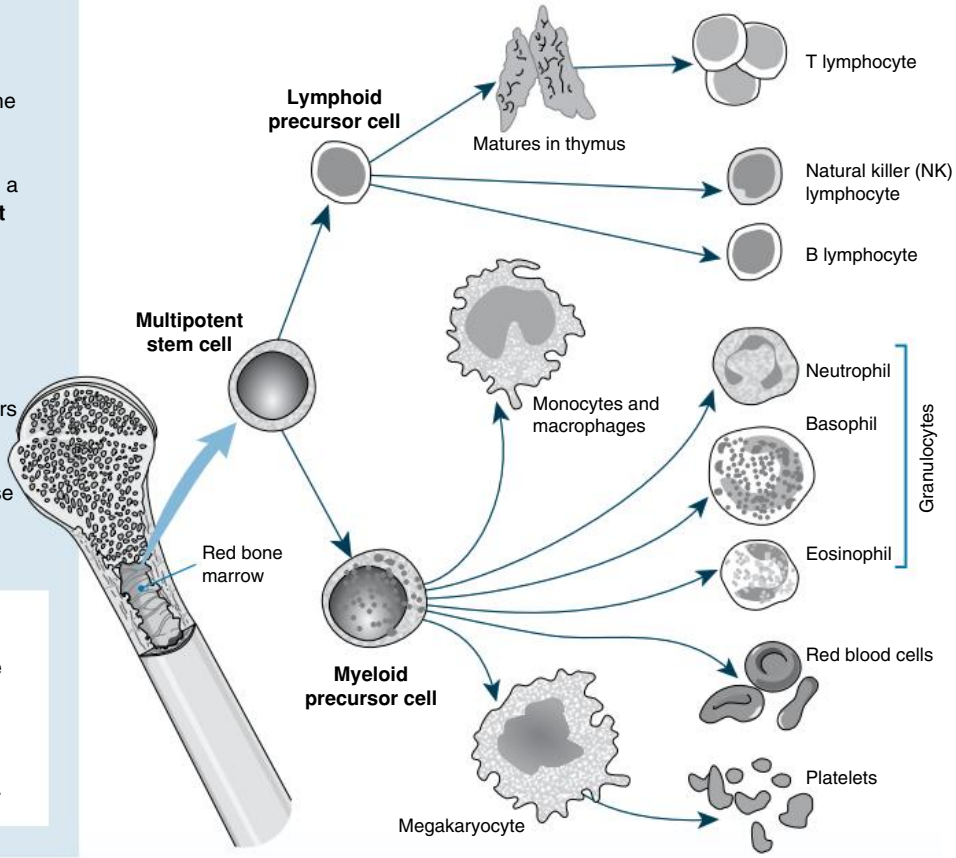
135 Stem Cells and Differentiation

Key Idea: Stem cells are undifferentiated cells found in multicellular organisms. They are characterised by the properties of self renewal and potency.

A zygote can differentiate into many different types of cells because early on it divides into stem cells. Stem cells are unspecialised and can give rise to the many cell types that make up the tissues and organs of a multicellular organism. The differentiation of multipotent stem cells in bone marrow gives rise to all the cell types that make up blood, a fluid connective tissue. Multipotent (or adult) stem cells are found in most body organs, where they replace old or damaged cells and replenish the body's cells throughout life.

Stem Cells and Blood Cell Production

New blood cells are produced in the red bone marrow, which becomes the main site of blood production after birth, taking over from the fetal liver. All types of blood cells develop from a single cell type: called a **multipotent stem cell**. These cells are capable of mitosis and of differentiation into 'committed' precursors of each of the main types of blood cell. Each of the different cell lines is controlled by a specific **growth factor**. When a stem cell divides, one of its daughters remains a stem cell, while the other becomes a precursor cell, either a **lymphoid cell** or **myeloid cell**. These cells continue to mature into the various specialised cell types.



Properties of Stem Cells

Self renewal: The ability to divide many times while maintaining an unspecialised state.

Potency: The ability to differentiate into specialised cells.

Categories of Stem Cells

Totipotent stem cells

These stem cells can differentiate into all the cells in an organism. Example: In humans, the zygote and its first few divisions. The meristematic tissue of plants is also totipotent.

Pluripotent stem cells

These stem cells can give rise to any cells of the body, except extra-embryonic cells (e.g. placenta and chorion). Example: Embryonic stem cells.

Multipotent stem cells

These adult stem cells can give rise a limited number of cell types, related to their tissue of origin. Example: Bone marrow stem cells, epithelial stem cells, bone stem cells (osteoblasts).

1. Describe the two defining features of stem cells:

   (a) _____

   (b) _____

2. Explain the role of stem cells in the development of specialised tissues in multicellular organisms: _____

_____

_____

_____

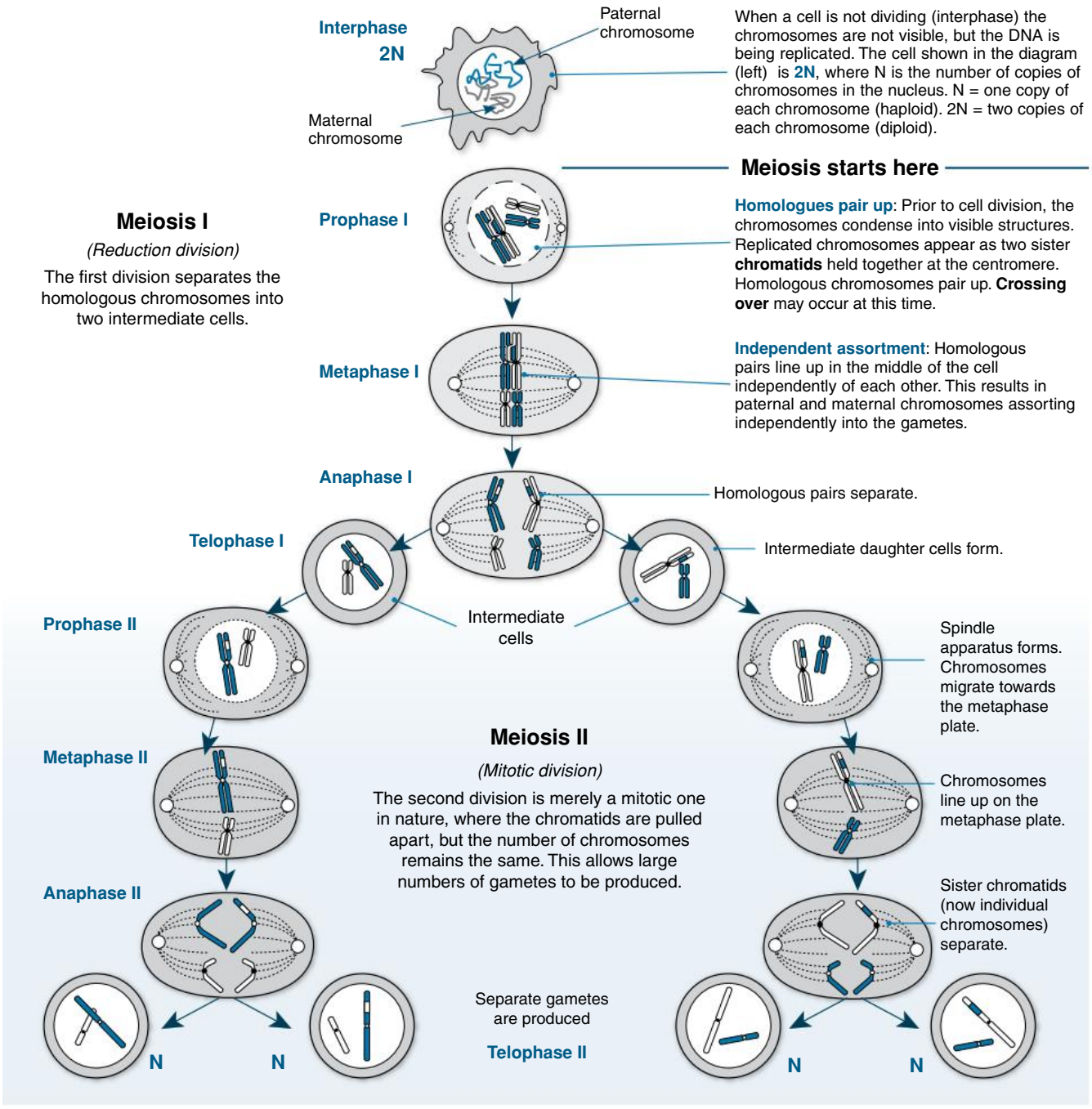
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KNOW LINK 163 LINK 280 LINK 281

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136 Meiosis

Key Idea: Meiosis is a special type of cell division. It produces sex cells (gametes) for the purpose of sexual reproduction. Meiosis involves a single chromosomal duplication followed by two successive nuclear divisions, and results in a halving of the diploid chromosome number. Meiosis occurs in the sex organs of plants and animals. If genetic mistakes (**gene** and **chromosome mutations**) occur here, they will be passed on to the offspring (they will be inherited).



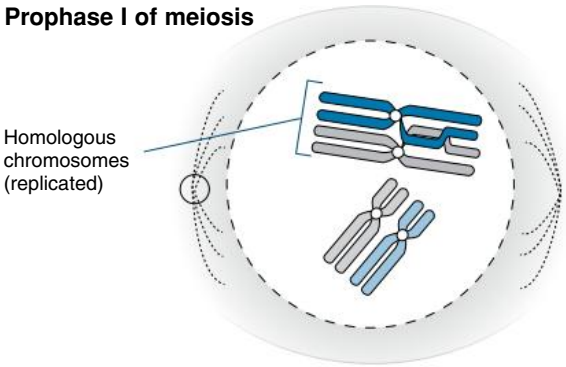
1. Describe the behaviour of the chromosomes in the first division of meiosis: _____

2. Describe the behaviour of the chromosomes in the second division of meiosis: _____

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Crossing Over and Recombination

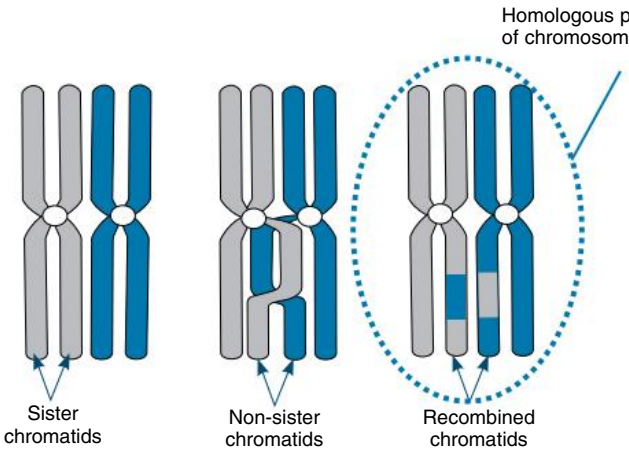
Prophase I of meiosis



In prophase I homologous chromosomes pair up to form **bivalents**. This process is called **synapsis**. While they are paired, the non-sister chromatids of homologous pairs may become tangled and segments may be exchanged in a process called **crossing over**. The crossing over occurs at points called chiasmata.

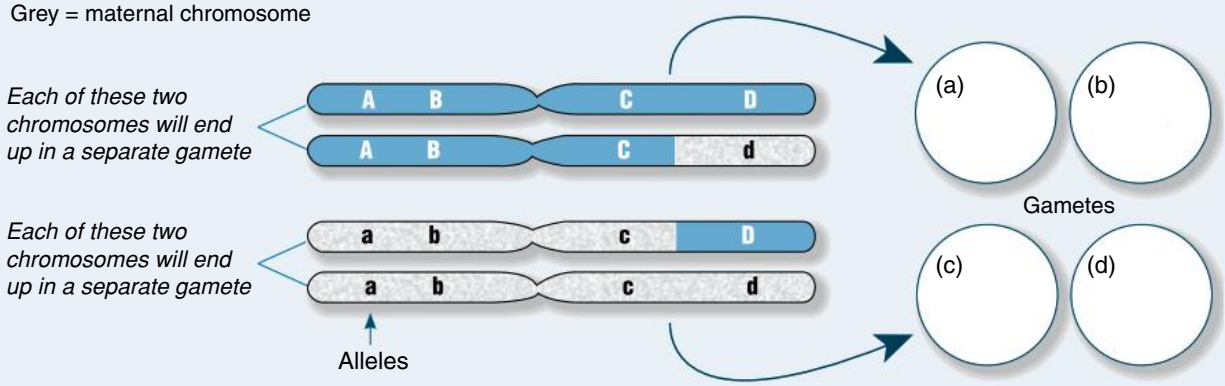
Crossing over results in the **recombination** of alleles, producing greater variation in the offspring than would otherwise occur.

Crossing over refers to the mutual exchange of pieces of chromosome (and their genes) between the **homologous** chromosomes. It can occur only during prophase in the first division of meiosis. Recombination as a result of crossing over is an important mechanism to increase genetic variability in the offspring and has the general effect of allowing genes to move independently of each other through the generations in a way that allows concentration of beneficial alleles.



Gamete Formation

Blue = paternal chromosome
Grey = maternal chromosome



The homologous chromosomes above have completed a crossing over event. Recombination have resulted in four different allele combinations instead the two that would appear if there was no crossing over.

3. (a) When does DNA replication occur? _____

   (b) What is the difference between a chromosome and a chromatid: _____

   _____

4. (a) Distinguish between a haploid and a diploid cell: _____

   _____

   _____

   (b) Circle the **haploid** cells in the diagram on the previous page

5. Complete the diagram above (a) - (d) by drawing the gametes formed:

6. (a) How does crossing over alter the genotype of the gametes produced by meiosis? _____

   _____

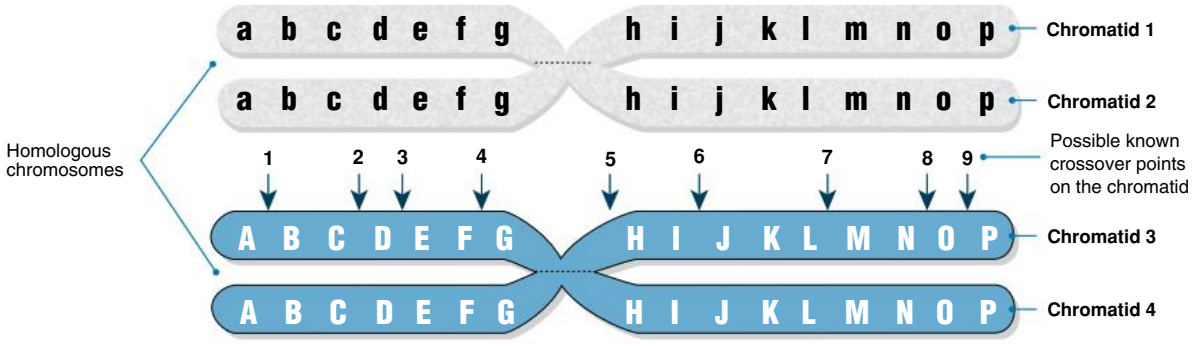
   (b) What is the consequence of this? _____

   _____

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137 Crossing Over Problems

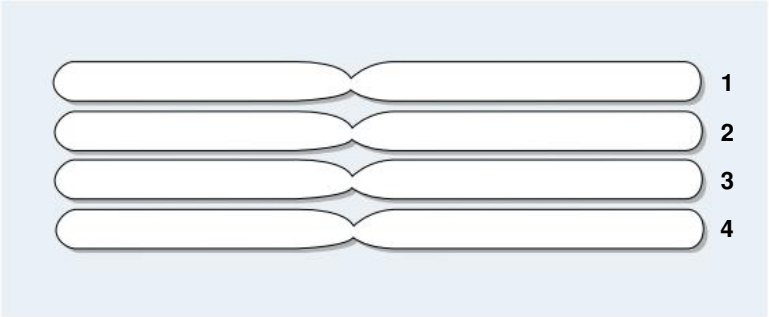
Key Idea: Crossing over can occur in multiple places in chromosomes, producing a huge amount of genetic variation. The diagram below shows a pair of homologous chromosomes about to undergo chiasma formation during the first cell division in the process of meiosis. There are known crossover points along the length of the chromatids (same on all four chromatids shown in the diagram). In the prepared spaces below, draw the gene sequences after crossing over has occurred on three unrelated and separate occasions (it would be useful to use different coloured pens to represent the genes from the two different chromosomes). See the diagrams on the previous page as a guide.



1. Crossing over occurs at a **single** point between the chromosomes above.

   (a) Draw the gene sequences for the four chromatids (on the right), after crossing over has occurred at crossover point: **2**

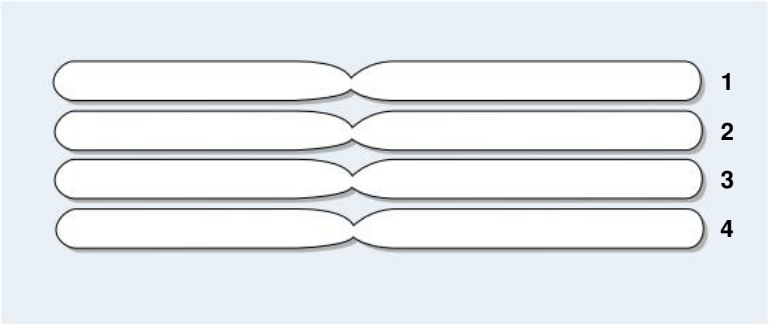
   (b) Which genes have been exchanged with those on its homologue (neighbour chromosome)?



2. Crossing over occurs at **two** points between the chromosomes above.

   (a) Draw the gene sequences for the four chromatids (on the right), after crossing over has occurred between crossover points: **6** and **7**.

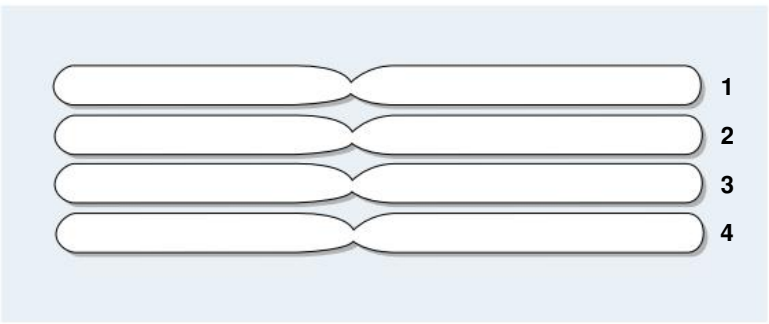
   (b) Which genes have been exchanged with those on its homologue (neighbour chromosome)?



3. Crossing over occurs at **four** points between the chromosomes above.

   (a) Draw the gene sequences for the four chromatids (on the right), after crossing over has occurred between crossover points: **1** and **3**, and **5** and **7**.

   (b) Which genes have been exchanged with those on its homologue (neighbour chromosome)?



4. What would be the genetic consequences if there was no crossing over between chromatids during meiosis?

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LINK 191 LINK 136 KNOW

138 Modelling Meiosis

Key Idea: We can simulate crossing over, gamete production, and the inheritance of alleles during meiosis using ice-block sticks to represent chromosomes.

This practical activity simulates the production of gametes (sperm and eggs) by **meiosis** and shows you how **crossing over** increases genetic variability. This is demonstrated by studying how two of your own alleles are inherited by the child produced at the completion of the activity. Completing this activity will help you to visualise and understand meiosis. It will take 25-45 minutes.

Background

Each of your somatic cells contain 46 chromosomes. You received 23 chromosomes from your mother (**maternal chromosomes**), and 23 chromosomes from your father (**paternal chromosomes**). Therefore, you have 23 homologous (same) pairs. For simplicity, the number of chromosomes studied in this exercise has been reduced to four (two homologous pairs). To study the effect of crossing over on genetic variability, you will look at the inheritance of two of your own traits: the ability to **tongue roll** and **handedness**.

Chromosome #	Phenotype	Genotype
10	Tongue roller	TT, Tt
10	Non-tongue roller	tt
2	Right handed	RR, Rr
2	Left handed	rr









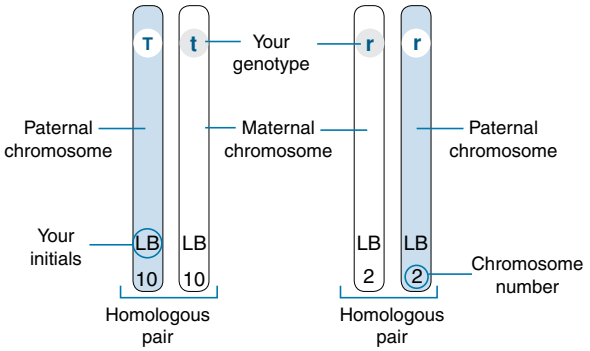
Trait	Phenotype	Genotype
Handedness		
Tongue rolling		

Record your phenotype and genotype for each trait in the table (right). **NOTE:** If you have a dominant trait, you will not know if you are heterozygous or homozygous for that trait, so you can choose either genotype for this activity.

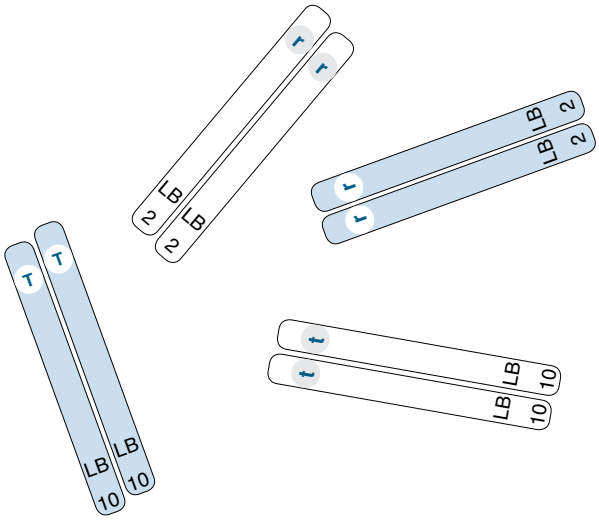
BEFORE YOU START THE SIMULATION: Partner up with a classmate. Your gametes will combine with theirs (fertilisation) at the end of the activity to produce a child. Decide who will be the female, and who will be the male. You will need to work with this person again at step 6.

1. Collect four ice-blocks sticks. These represent four chromosomes. Colour two sticks blue or mark them with a P. These are the paternal chromosomes. The plain sticks are the maternal chromosomes. Write your initial on each of the four sticks. Label each chromosome with their chromosome number (right).

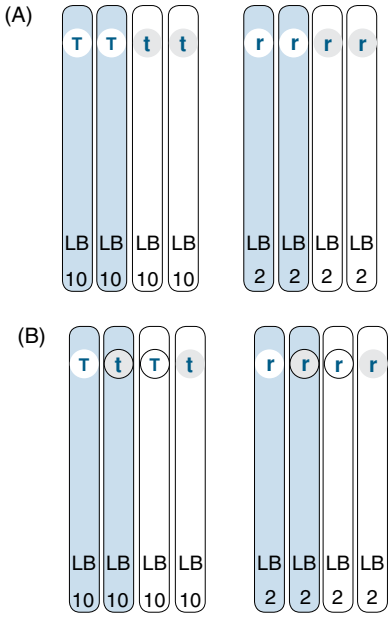
   Label four sticky dots with the alleles for each of your phenotypic traits, and stick it onto the appropriate chromosome. For example, if you are heterozygous for tongue rolling, the sticky dots with have the alleles **T** and **t**, and they will be placed on chromosome 10. If you are left handed, the alleles will be **r** and **r** and be placed on chromosome 2 (right).



2. Randomly drop the chromosomes onto a table. This represents a cell in either the testes or ovaries. **Duplicate** your chromosomes (to simulate DNA replication) by adding four more identical ice-block sticks to the table (below). This represents **interphase**.

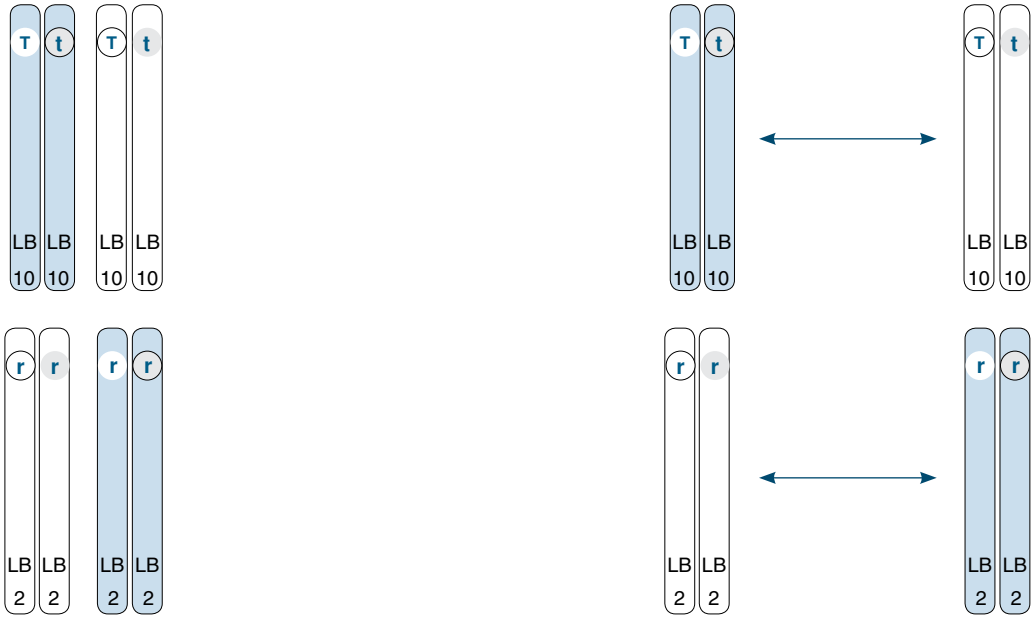


3. Simulate **prophase I** by lining the duplicated chromosome pair with their homologous pair (below). For each chromosome number, you will have four sticks touching side-by-side (A). At this stage **crossing over** occurs. Simulate this by swapping sticky dots from adjoining homologs (B).



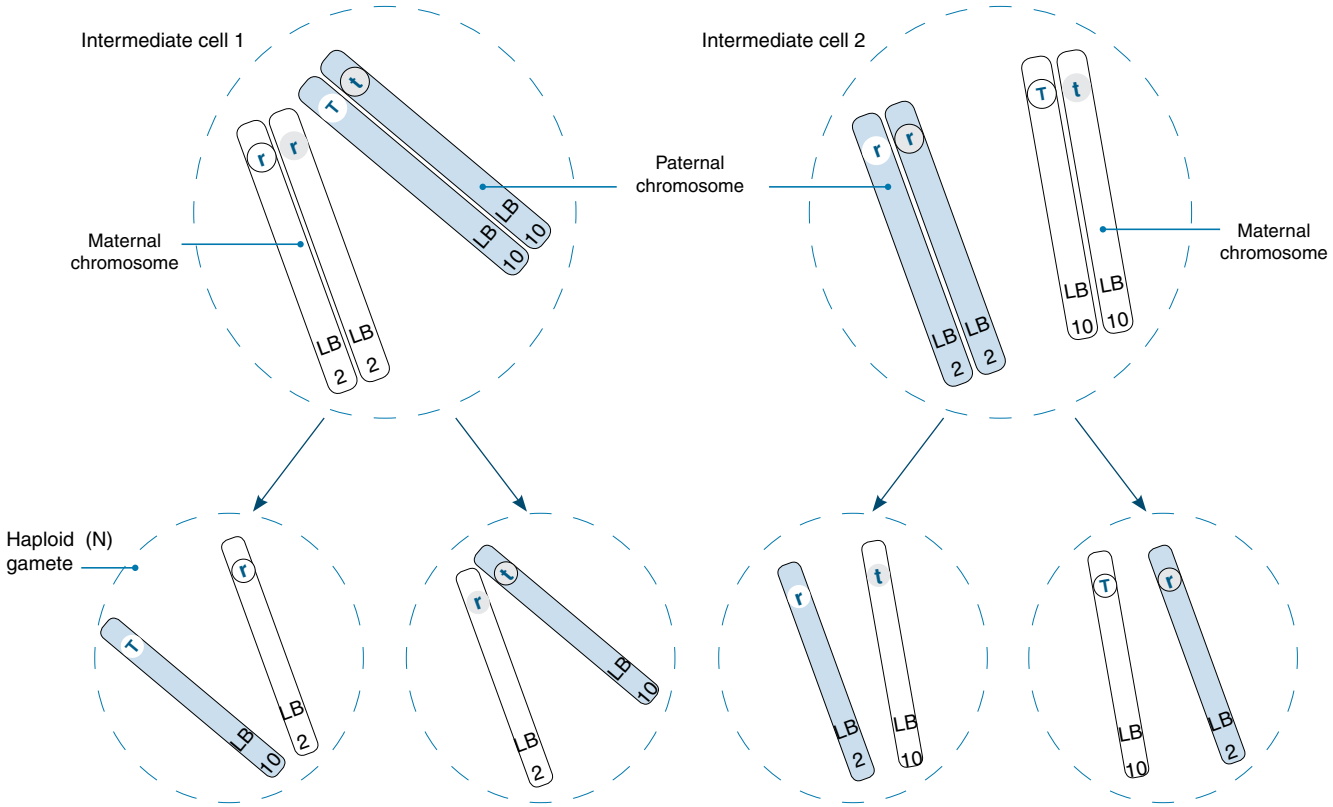
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4. Randomly align the homologous chromosome pairs to simulate alignment on the metaphase plate (as occurs in **metaphase I**). Simulate **anaphase I** by separating chromosome pairs. For each group of four sticks, two are pulled to each pole.



5. **Telophase I:** Two intermediate cells are formed. If you have been random in the previous step, each intermediate cell will contain a mixture of maternal and paternal chromosomes. This is the end of **meiosis 1**.

   Now that meiosis 1 is completed, your cells need to undergo **meiosis 2.** Carry out prophase II, metaphase II, anaphase II, and telophase II. Remember, there is no crossing over in meiosis II. At the end of the process each intermediate cell will have produced two haploid gametes (below).



6. Pair up with the partner you chose at the beginning of the exercise to carry out **fertilisation**. Randomly select one sperm and one egg cell. The unsuccessful gametes can be removed from the table. Combine the chromosomes of the successful gametes. You have created a child! Fill in the following chart to describe your child's genotype and phenotype for tongue rolling and handedness.

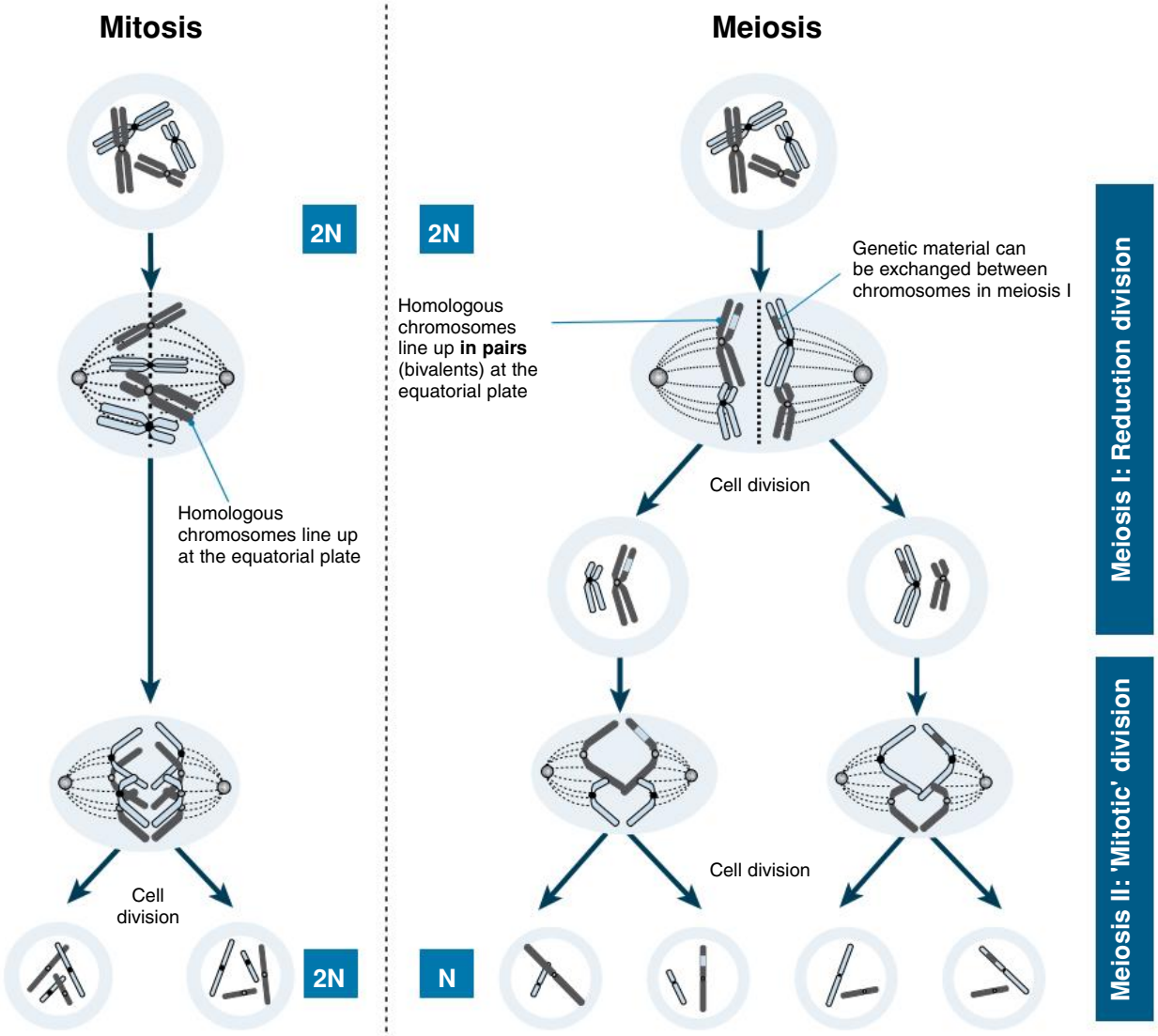
Trait	Phenotype	Genotype
Handedness		
Tongue rolling		

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139 Mitosis vs Meiosis

Key Idea: Mitosis produces two daughter cells genetically identical to the parent cell. Meiosis produces four daughter cells that contain half the genetic information of the parent cell.

Cell division is fundamental to all life, as cells arise only by the division of existing cells. All types of cell division begin with replication of the cell's DNA. In eukaryotes, this is followed by division of the nucleus. There are two forms of nuclear division: **mitosis** and **meiosis**, and they have quite different purposes and outcomes. Mitosis is the simpler of the two and produces two identical daughter cells from each parent cell. Mitosis is responsible for growth and repair processes in multicellular organisms and reproduction in single-celled and asexual eukaryotes. Meiosis involves a **reduction division** in which haploid gametes are produced for the purposes of sexual reproduction. Fusion of haploid gametes in fertilisation restores the diploid cell number in the **zygote**.



1. Explain how mitosis conserves chromosome number while meiosis reduces the number from diploid to haploid:

2. Describe a fundamental difference between the first and second divisions of meiosis:

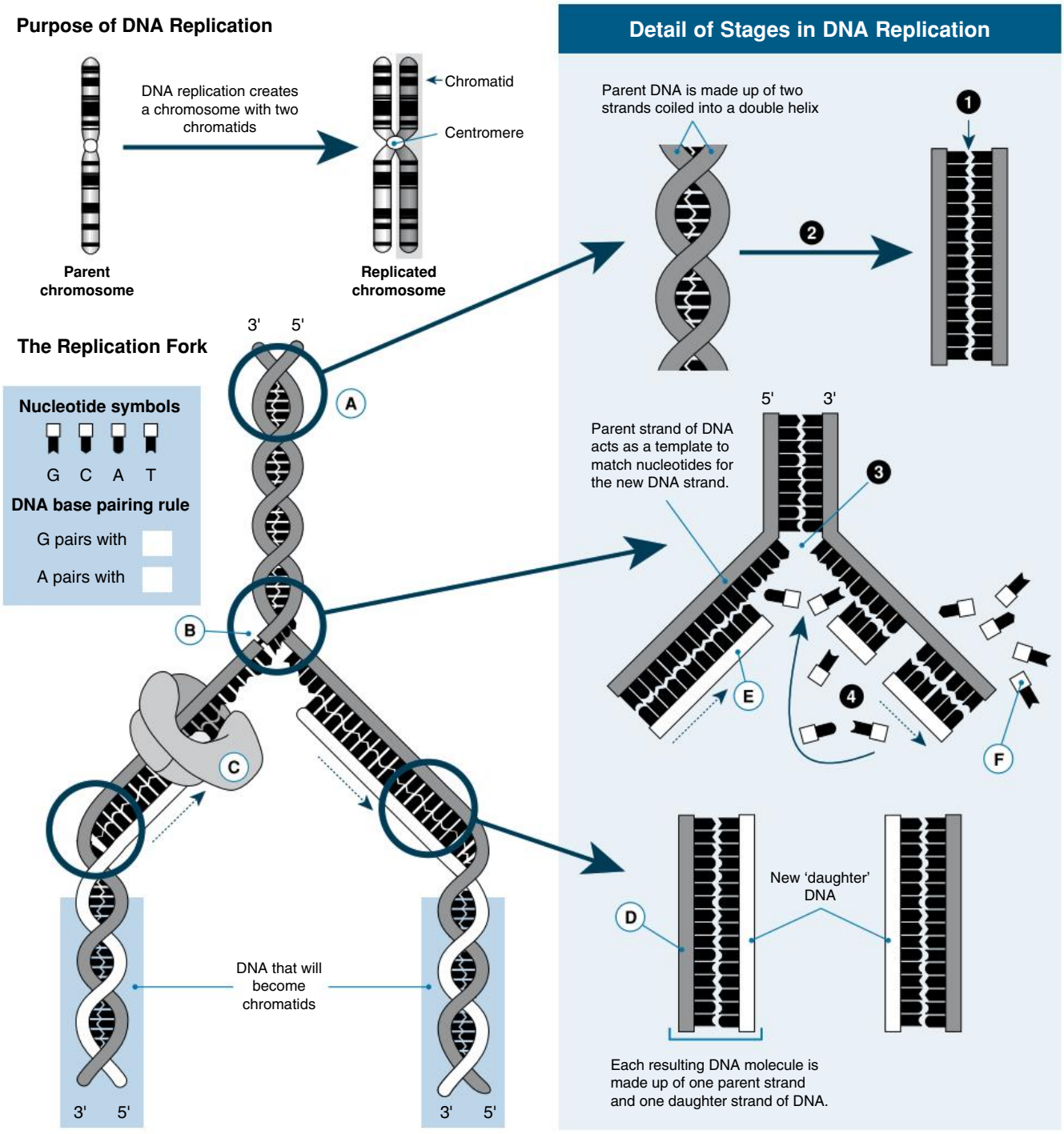
3. How does meiosis introduce genetic variability into gametes and offspring (following gamete fusion in fertilisation)?

KNOW LINK 133 LINK 136

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140 KEY TERMS: Did You Get It?

The diagram below is a simplified summary of the main steps in DNA replication. You should use this activity to test your understanding of the main features of DNA replication, using the knowledge gained in the earlier activity to fill in the missing information. You should attempt this from what you have learned, but refer to the earlier activity if necessary.



1. In the white boxes provided in the diagram, state the base pairing rule for making a strand of DNA:

2. Identify each of the structures marked with a letter. (A-F):

   A: _____ C: _____ E: _____

   B: _____ D: _____ F: _____

3. Match each of the processes (1-4) to the correct summary of the process provided below:

   ☐ Unwinding of parent DNA double helix

   ☐ Unzipping of parent DNA

   ☐ Free nucleotides occupy spaces alongside exposed bases

   ☐ DNA strands are joined by base pairing

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LINK 128 TEST

QLD As required
VCE Unit 4 (1)
WA ATAR Unit 3
SA Theme O

Gene Technology

Key terms

annealing
biotechnology
blunt end
cloning
DNA amplification
DNA (genetic) profiling
DNA ligase
DNA ligation
DNA polymerase
DNA sequencing
forensics
gel electrophoresis
gene technology
genetic modification
genome projects
GMO
marker gene
microsatellite
molecular clone
plasmid
polymerase chain reaction
primer
recognition site
recombinant DNA technology
restriction digestion
restriction enzyme
reverse transcription
sticky end
transgenic organism
transgenesis
vector

Basic principles of biotechnology

Learning aims and skills

Activity number

- ☐ 1 Describe the scope of biotechnology, distinguishing it from gene technology, but recognising the connections between the two. Recognise that the same basic techniques (#2-4) are used in a range of different processes and applications. — 141-143
- ☐ 2 Explain the use of restriction enzymes in recombinant DNA technology, distinguish between sticky-end and blunt-end fragments. Describe the technique and purpose of DNA ligation and annealing, including the role of DNA ligase. — 144
- ☐ 3 Explain the role of gel electrophoresis (of DNA) in gene technology. Explain how the DNA fragments on a gel are made visible and the role of DNA markers in identifying fragments of different size. — 145 146
- ☐ 4 Describe and explain the role of polymerase chain reaction (PCR) in DNA amplification, including the role of primers and DNA polymerase. — 147 148





Processes and applications

Learning aims and skills

Activity number

- ☐ 5 PROFILING: Describe and explain DNA profiling using PCR. Discuss the applications of DNA profiling as a forensic and diagnostic tool. — 149-151
- ☐ 6 SEQUENCING: Describe and explain the use of PCR and gel electrophoresis in DNA sequencing. Explain the role of automated sequencing in the feasibility of large scale genome analyses and discuss current and future applications of sequencing technology. — 152-154
- ☐ 7 *IN-VIVO* GENE CLONING: Describe and explain gene cloning using plasmids. Discuss one or more of the applications of gene cloning. — 155 156
- ☐ 8 TRANSGENESIS: Describe and explain transgenesis, including the role of vectors in integrating foreign DNA into another genome. Describe the applications of transgenic organisms, and discuss any ethical concerns associated with their production and use. — 143 144 157-161
- ☐ 9 CASE STUDIES IN BIOTECHNOLOGY: Describe and explain how gene technology is used to meet human needs or demands in the areas of medicine, forensics, agriculture, or phylogenetics. Examples could include enzyme production, crop improvement, protein production, or gene therapy. — 150 151 152 154 157-160 280-282
- ☐ 10 MEDICAL BIOTECHNOLOGY: Not all medical applications of biotechnology involve genetic modification. Describe and explain the use of monoclonal antibodies, tissue engineering, organ transplants, and stem cell technology. Discuss any ethical concerns with these applications. — 72 104 105 283 284



141 A Brief History of Human Genetic Research

Key Idea: Humans have been manipulating the transfer of genetic information between organisms ever since plants and animals were first domesticated over 10,000 years ago.

Selective breeding, the breeding together of organisms with desirable traits, was the earliest form of genetic manipulation. In the last few decades, humans have developed sophisticated methods to manipulate genetic information to produce organisms with beneficial traits. For example, the insertion of a gene from one species into another completely different species, is now common-place. The time-line below outlines some significant steps in the history of humans manipulating the transfer of genetic information.

~8,000 BC

Cultivation of grains and domestication of animals.



Teosinte, a wild maize.

John Doebley

~3,000 BC

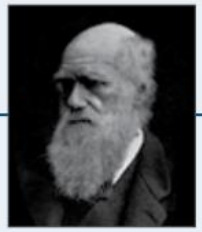


Mules created by crossing donkeys and horses.

3,000 BC-AD 1800

Breeds of plants and animals refined. Many of today's common varieties and breeds appeared by mid 1800s.





AD 1850

1859 Charles Darwin publishes Origin of Species in which he details the selective breeding of pigeons to produce breeds with radically different appearances.

1890 Rabbits are born from embryos that were implanted in surrogate mothers.



AD 1920

1926 Hybridisation of plant crops produces higher yielding seeds.

1930 First collection of bovine embryos for transfer to surrogate mothers.

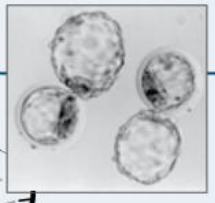
1940 Mutagens used on plants to alter genes and produce new plant varieties.



AD 1950

1951 First successful transfer of bovine embryos to surrogate mothers.

1953 Structure of DNA proposed by Watson and Crick.





PCR machine

AD 1970

1972 First *in vitro* recombinant DNA. Frozen mouse embryo revived, implanted, and born.

1973 First transgenic bacteria produced.

1976 Prenatal genetic diagnosis using DNA.

1978 First human baby from *in vitro* fertilisation born.

1979 Embryo splitting to produce twin lambs. Insulin produced by GM bacteria.

AD 1980-1990

Achievements include: *In vitro* fertilisation of bovine ovum, transgenic mice, PCR, transgenic pigs, first transgenic plant resistant to insects, location of gene for Huntington's disease, transgenic maize, transgenic plants for pharmaceuticals.



Flavr savr tomato

AD 1991-2000

Achievements include: gene therapy, plant IVF, Flavr savr GE tomato commercially available, cloning produces Dolly the sheep, completion of first draft of human genome.



AD 2001 onwards

Achievements include: First gene therapy successes (SCID), genetically modified rhesus monkey, mouse with two female parents born, various cloned animals produced, Glofish - the first genetically engineered pets commercially available.

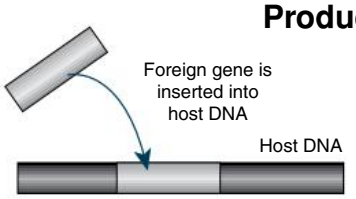
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142 What is Genetic Modification?

Key Idea: Genetically modified organisms (GMOs) are organisms with artificially altered DNA. GMOs may provide solutions to many human health and food supply issues.

The genetic modification of organisms has many potential applications including medical cures, increasing crop yields, and potentially helping to solve the world's pollution and resource problems. Organisms with artificially altered DNA are referred to as **genetically modified organisms** or **GMOs**. They may be modified in one of three ways (see below). Some of the current and proposed applications of gene technology raise complex ethical and safety issues. The benefits of their use must be carefully weighed against the risks to human health, and the health and well-being of other organisms and the environment.

Producing Genetically Modified Organisms (GMOs)

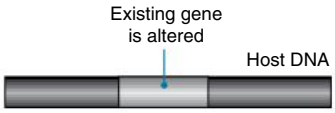


Add a foreign gene

A novel (foreign) gene is inserted from another species. This will enable the GMO to express the trait coded by the new gene. Organisms genetically altered in this way are referred to as **transgenic.**



Human insulin, used to treat diabetic patients, is now produced using transgenic bacteria.



Alter an existing gene

An existing gene may be altered to make it express at a higher level (e.g. growth hormone) or in a different way (in tissue that would not normally express it). This method is also used for gene therapy.



Gene therapy could be used treat genetic disorders, such as cystic fibrosis.



Delete or 'turn off' a gene

An existing gene may be deleted or deactivated (switched off) to prevent the expression of a trait (e.g. the deactivation of the ripening gene in tomatoes produced the Flavr-Savr tomato).



Manipulating gene action is one way in which to control processes such as ripening in fruit.

1. Using examples, discuss the ways in which an organism may be genetically modified (to produce a GMO):

2. Explain how human needs or desires have provided a stimulus for the development of the following biotechnologies:

   (a) Gene therapy:

   (b) The production and use of transgenic organisms:

   (c) Plant micropropagation (tissue culture):

KNOW LINK 141 LINK 158 LINK 159 LINK 279

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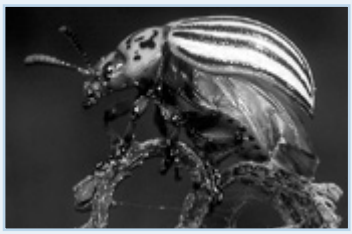
143 Applications of GMOs

Key Idea: GMOs are used in the food industry, agriculture, horticulture, medicine, and environmental practices.

Techniques for genetic manipulation are now widely applied throughout modern biotechnology: in food and enzyme technology, in industry and medicine, and in agriculture and horticulture. Microorganisms are among the most widely used GMOs, with applications ranging from pharmaceutical production and vaccine development to environmental clean-up and rehabilitation. Crop plants are also commonly genetically modification because they are easily propagated and the potential gains are great. However their use is controversial because transgenes are easily spread between plant species.



Extending shelf life: Shelf life in fresh produce (e.g. tomatoes) can be extended by switching off the genes for specific enzymes involved in the fruit ripening process (e.g. the enzymes involved in softening of the fruit wall or controlling the production of ethylene).



Pest or herbicide resistance: Plants can be engineered to carry and express genes for insect toxins or herbicide resistance. Pest resistant crops do not require spraying and herbicide resistance allows the grower to control weeds without damaging the crops.



Biofactories: Transgenic bacteria are widely used to produce desirable commodities, such as hormones or proteins. Large quantities of a product can be produced using bioreactors. One example includes injectable human insulin produced by recombinant bacteria or yeast (above).



Vaccine development: Multipurpose vaccines can be made using gene technology. Genes encoding antigenic vaccine components (e.g. viral protein coat) are inserted into an unrelated live vaccine (e.g. polio vaccine) and deliver proteins to stimulate an immune response in the recipient.



Crop improvement: Gene technology is now an integral part of the development of new crop varieties. Crops can be engineered to produce higher protein or vitamin levels (e.g. golden rice) or to grow in inhospitable conditions (e.g. salty or arid land).



Environmental clean-up: Bacteria can be engineered to thrive on waste products, such as liquefied newspaper pulp or oil. They degrade pollutants and wastes, and minerals (e.g. mercury) may also be recovered from the bacteria after the cleanup.





Livestock improvement using transgenic animals: Transgenic sheep have been used to enhance wool production in flocks (above, left). The keratin protein of wool is largely made of a single amino acid, cysteine. Injecting developing sheep with the genes for the enzymes that generate cysteine produces woollier sheep. Transgenic sheep carrying the human gene for a protein, α-1-antitrypsin, produce the protein in their milk. The antitrypsin is extracted from the milk and can be used to treat hereditary emphysema.

1. Suggest one economic advantage of extending shelf life in fresh produce:

2. Suggest one disadvantage of engineering crop plants to be herbicide resistant:

3. What is the advantage of using GE bacteria to produce a human hormone like insulin?

4. Why might it be safer to produce a vaccine using gene technology, rather than the pathogen itself?

5. What advantages could be gained by developing a GE crop that produces more protein?

6. Using animals as live biofactories to produce a valuable product (e.g. a human protein) is controversial. What welfare issues could be associated with such practices?

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LINK 161 LINK 159 LINK 157 LINK 103 KNOW

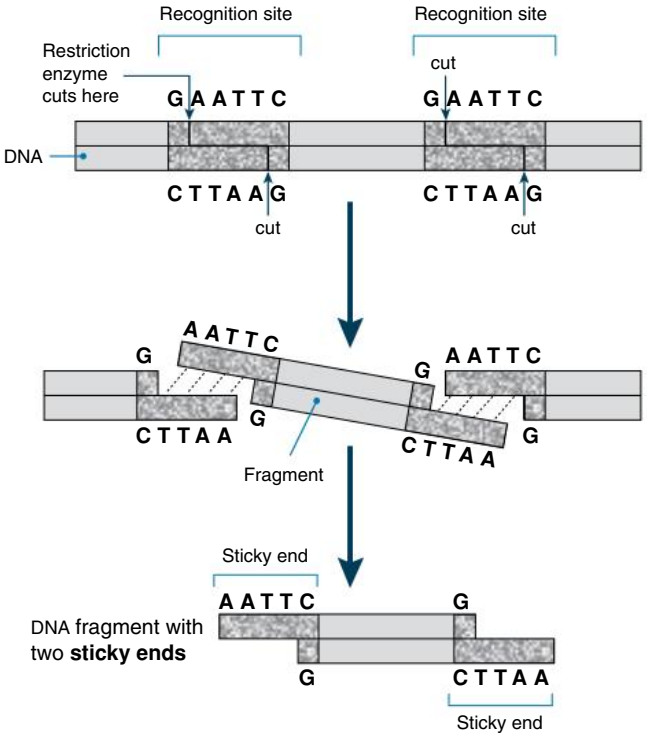
144 Making Recombinant DNA

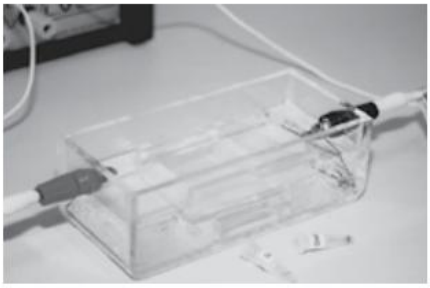
Key Idea: Recombinant DNA (rDNA) is produced by first isolating a DNA sequence, then inserting it into the DNA of a different organism.

The production of rDNA is possible because the DNA of every organism is made of the same building blocks (**nucleotides**). rDNA allows a gene from one organism to be moved into, and expressed in, a different organism. Two important tools used to create rDNA are restriction digestion (chopping up the DNA) using **restriction enzymes** and DNA ligation (joining of sections of DNA) using the enzyme **DNA ligase**.

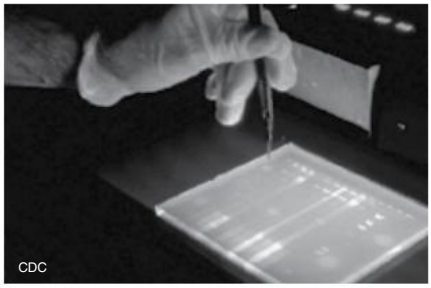
Information about Restriction Enzymes

1. A **restriction enzyme** is an enzyme that cuts a double-stranded DNA molecule at a specific **recognition site** (a specific DNA sequence). There are many different types of restriction enzymes, each has a unique recognition site.
2. Some restriction enzymes produce DNA fragments with two **sticky ends** (right). A sticky end has exposed nucleotide bases at each end. DNA cut in such a way is able to be joined to other DNA with matching sticky ends. Such joins are specific to their recognition sites.
3. Some restriction enzymes produce a DNA fragment with two **blunt ends** (ends with no exposed nucleotide bases). The piece it is removed from is also left with blunt ends. DNA cut in such a way can be joined to any other blunt end fragment. Unlike sticky ends, blunt end joins are non-specific because there are no sticky ends to act as specific recognition sites.





The fragments of DNA produced by the restriction enzymes are mixed with ethidium bromide, a molecule that fluoresces under UV light. The DNA fragments are then placed on an electrophoresis gel to separate the different lengths of DNA.



Once the DNA fragments are separated, the gel is placed on a UV viewing platform. The area of the gel containing the DNA fragments of the correct length is cut out and placed in a solution that dissolves the gel. This releases the DNA into the solution.



The solution containing the DNA is centrifuged at high speed to separate out the DNA. Centrifugation works by separating molecules of different densities. Once isolated, the DNA can be spliced into another DNA molecule.

1. What is the purpose of restriction enzymes in making recombinant DNA? _____

2. Distinguish between sticky end and blunt end fragments: _____

3. Why is it useful to have many different kinds of restriction enzymes? _____

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Creating a Recombinant DNA Plasmid

1. Two pieces of DNA are cut by the same restriction enzyme (they will produce fragments with matching **sticky ends**).

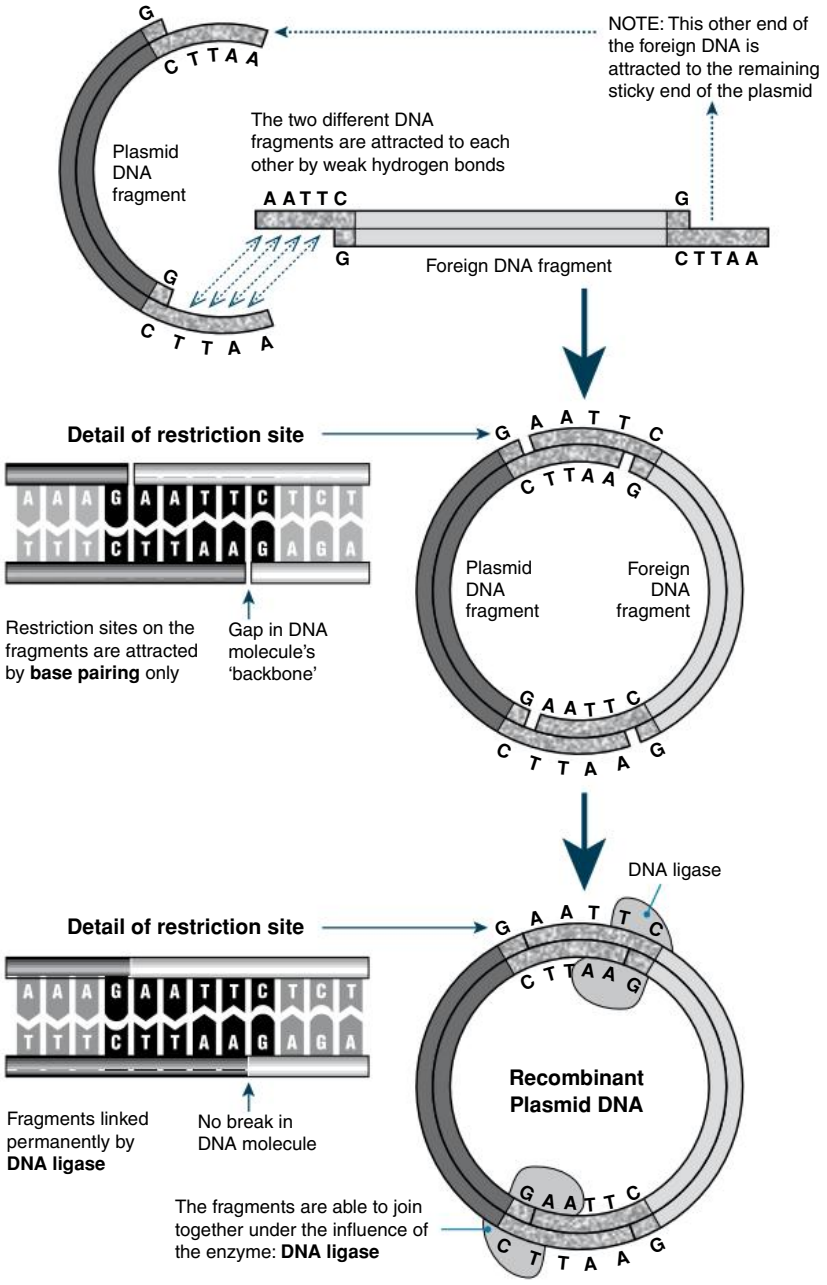
2. Fragments with matching sticky ends can be joined by base-pairing. This process is called **annealing.** This allows DNA fragments from different sources to be joined.

3. The fragments of DNA are joined together by the enzyme **DNA ligase**, producing a molecule of **recombinant DNA**.

4. The joined fragments will usually form either a linear or a circular molecule, as shown here (right) as recombinant **plasmid** DNA.



pGLO is a plasmid engineered to contain Green Fluorescent Protein (*gfp*). pGLO has been used to create fluorescent organisms, including the bacteria above (bright patches on agar plates).



4. Explain in your own words the two main steps in the process of joining two DNA fragments together:

   (a) Annealing: _____

   _____

   (b) DNA ligase: _____

   _____

5. Explain why **ligation** can be considered the reverse of the **restriction digestion** process:

   _____

6. Why can recombinant DNA be expressed in any kind of organism, even if it contains DNA from another species?

   _____

   _____

   _____

   _____

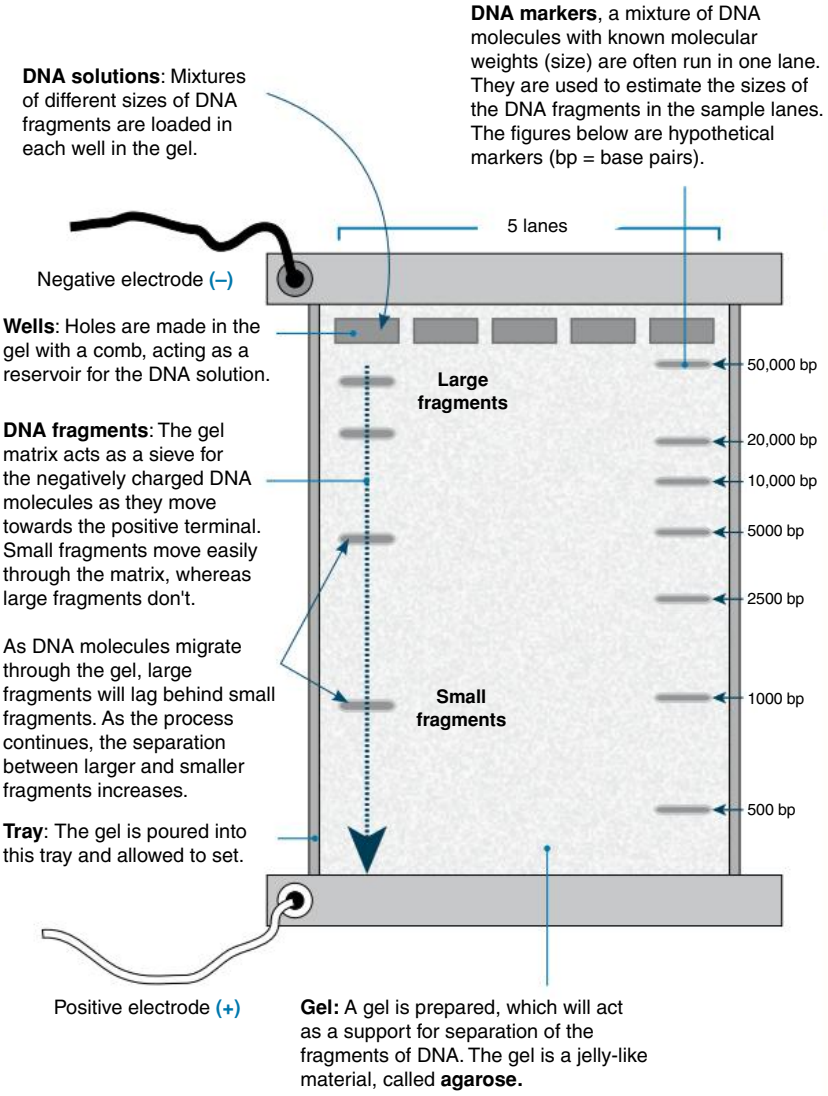
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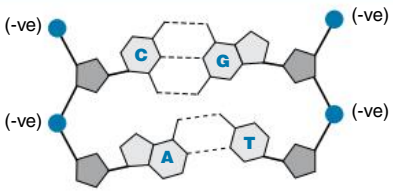
145 Gel Electrophoresis

Key Idea: Gel electrophoresis is used to separate DNA fragments on the basis of size.

DNA can be loaded onto an electrophoresis gel and separated by size. DNA has an overall negative charge, so when an electrical current is run through a gel, the DNA moves towards the positive electrode. The rate at which the DNA molecules move through the gel depends primarily on their size and the strength of the electric field. The gel they move through is full of pores (holes). Smaller DNA molecules move through the pores more quickly than larger ones. At the end of the process, the DNA molecules can be stained and visualised as a series of bands. Each band contains DNA molecules of a particular size. The bands furthest from the start of the gel contain the smallest DNA fragments.

Analysing DNA using Gel Electrophoresis





DNA is **negatively charged** because the phosphates (blue) that form part of the backbone of a DNA molecule have a negative charge.

Steps in the process of gel electrophoresis of DNA

1. A tray is prepared to hold the gel matrix.
2. A gel comb is used to create holes in the gel. The gel comb is placed in the tray.
3. Agarose gel powder is mixed with a buffer solution (this stabilises the DNA). The solution is heated until dissolved and poured into the tray and allowed to cool.
4. The gel tray is placed in an electrophoresis chamber and the chamber is filled with buffer, covering the gel. This allows the electric current from electrodes at either end of the gel to flow through the gel.
5. DNA samples are mixed with a "loading dye" to make the DNA sample visible. The dye also contains glycerol or sucrose to make the DNA sample heavy so that it will sink to the bottom of the well.
6. The gel is covered, electrodes are attached to a power supply and turned on.
7. When the dye marker has moved through the gel, the current is turned off and the gel is removed from the tray.
8. DNA molecules are made visible by staining the gel with **methylene blue** or ethidium bromide which binds to DNA and will fluoresce in UV light.

1. What is the purpose of gel electrophoresis? _____

_____

_____

2. Describe the two forces that control the speed at which fragments pass through the gel:

   (a) _____

   (b) _____

3. Why do the smallest fragments travel through the gel the fastest? _____

_____

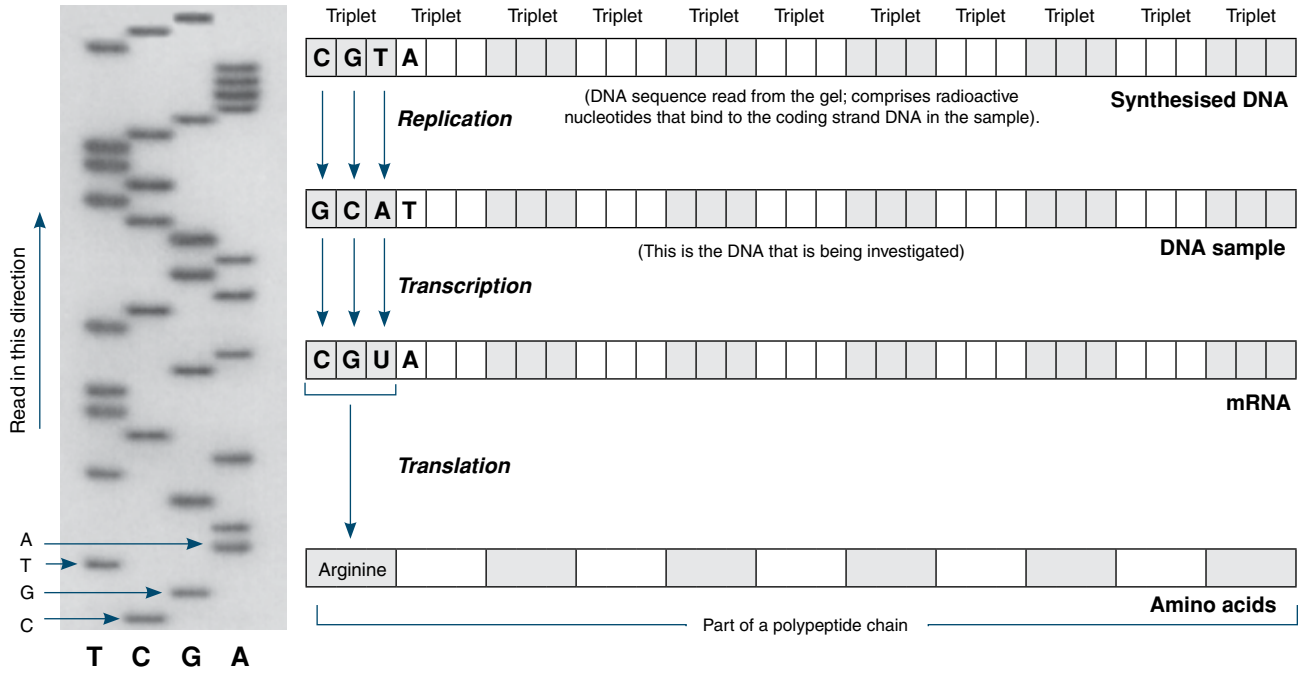
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146 Interpreting Electrophoresis Gels

Key Idea: The banding pattern on an electrophoresis gel can give information on DNA sequence and genetic relationships.

Once made, an electrophoresis gel must be interpreted. If a specific DNA base sequence was being investigated, then the band pattern can be used to determine the DNA sequence and the protein that it codes for. Alternatively, the banding pattern may be used as a profile for a species or individual. The pattern may be matched to an existing profile and identified or added to a database as a new DNA profile. Profiles can also be used to calculate relatedness based on the number of matching bands. From this information, a genetic tree (dendrogram) can be drawn.



1. Determine the amino acid sequence of a protein from the nucleotide sequence of its DNA, with the following steps:
   (a) Determine the sequence of **synthesised DNA** in the gel in the photograph above.
   (b) Convert it to the complementary sequence of the **sample DNA**.
   (c) Complete the **mRNA** sequence.
   (d) Determine the **amino acid** sequence by using the 'mRNA amino acid table' in this manual.

   NOTE: The nucleotides in the gel are read from bottom to top and the sequence is written in the spaces provided from left to right (the first 4 have been done for you).

2. For each single strand DNA sequence below, write the base sequence for the **complementary DNA** strand:

   (a) DNA: T A C T A G C C G C G A T T T A C A A T T

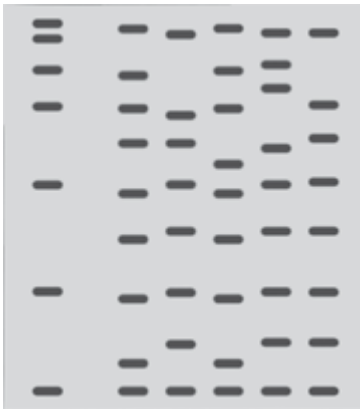
   DNA: _____

   (b) DNA: T A C G C C T T A A A G G G C C G A A T C

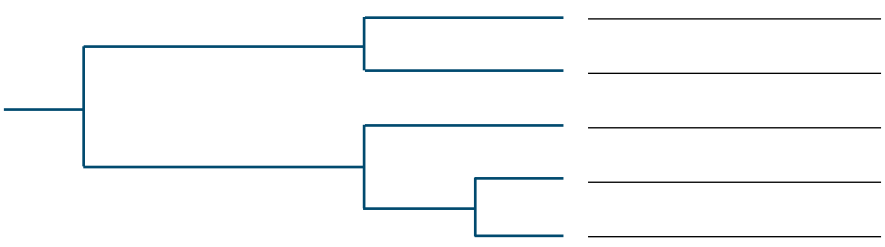
   DNA: _____

   (c) Which cell process does this exercise represent? _____

   _____



3. Determine the relatedness of each individual (A-E) using each banding pattern on the set of DNA profiles (left). When you have done this, complete the dendrogram by adding the letter of each individual.



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LINK 145 KNOW

147 Amazing Organisms, Amazing Enzymes

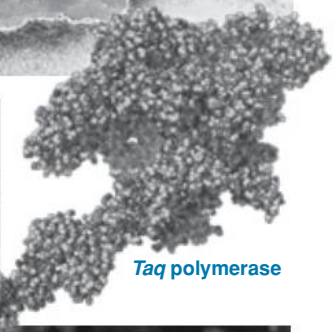
Key Idea: The substances produced by organisms to survive in their environment can provide solutions to biotechnological problems.

Before the 1980s scientists knew of only a few organisms that could survive in extreme conditions. Indeed, many scientists believed that life in highly saline or high temperature and pressure environments was impossible. That view changed with the discovery of bacteria inhabiting the deep sea hydrothermal vents. They tolerate temperatures over 110ºC and pressures of over 200 atmospheres. Bacteria were also found in volcanic hot pools on land, some surviving at temperatures in excess of 80ºC. Most enzymes are denatured at temperatures above 40ºC, but these **thermophilic** bacteria have enzymes that are fully functional at high temperatures. This discovery led to the development of one of the most important techniques in biotechnology, the **polymerase chain reaction** (PCR).

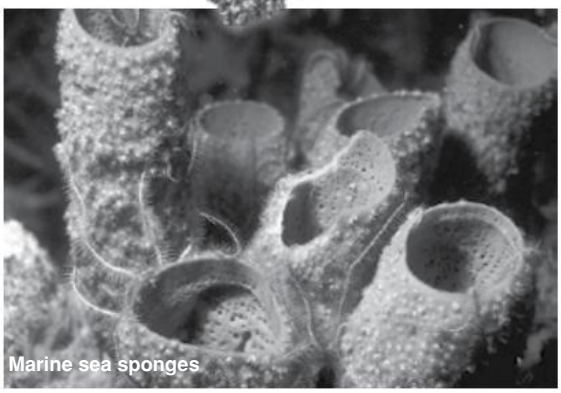

Hot springs

PCR is a technique, first described in the 1970s, that allows scientists to copy and multiply a piece of DNA millions of times. The DNA is heated to 98ºC so that it separates into single strands and polymerase enzyme is added to synthesise new DNA strands from supplied free nucleotides. This earlier technique was labor intensive and expensive because the polymerase denatured at the high temperatures and had to be replaced every cycle. In 1985, a thermophilic polymerase (***Taq* polymerase**) was isolated from the bacterium *Thermophilus aquaticus,* which inhabited the hot springs of Yellowstone National Park. Isolating this enzyme enabled automation of the PCR process, because the polymerase was stable throughout multiple cycles of synthesis. This led to a rapid growth in biotechnology, and gene technology in particular, because DNA samples could be easily copied for sequencing.


Thermophilus aquaticus


***Taq* polymerase**

Searching for novel compounds in organisms from extreme environments is important in the development of new biotechnologies. Organisms must have compounds that can work in their specific environment, and the identification and extraction of these may allow them to be adapted for human use. For example, the Antarctic sea sponge *Kirkpatrickia variolosa* produces an alkaloid excreted as a toxic defence to prevent other organisms growing nearby. Tests indicate that this same chemical may have biological activity against cancer cells. Compounds from other sponge species are currently being assessed to treat a range of diseases including cancer, AIDS, tuberculosis and other bacterial infections, and cystic fibrosis.


Marine sea sponges

1. Why was PCR not a viable technique until the mid 1980s? _____

2. Explain why *Taq* polymerase was so important in the development of PCR: _____

3. Discuss how investigating the ecology and physiology of organisms can lead to advances in unrelated areas of science:

LINK
COMP 148

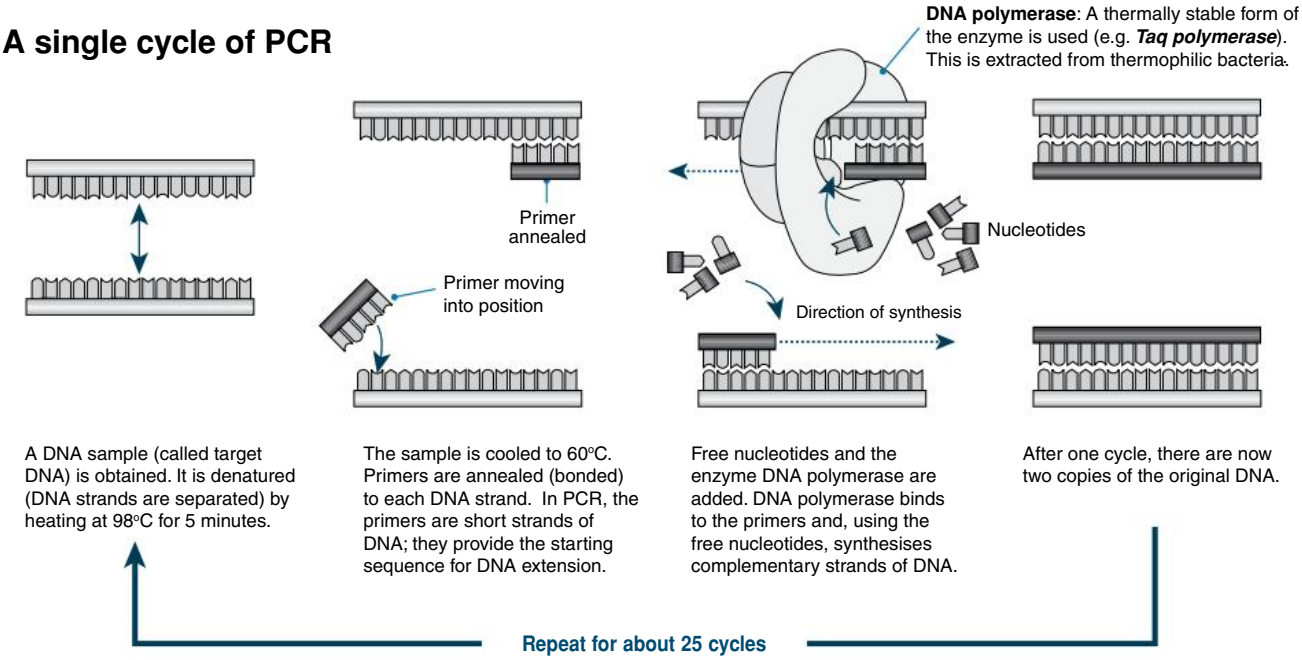
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148 DNA Amplification Using PCR

Key Idea: PCR uses a polymerase enzyme to copy a DNA sample, producing billions of copies in a few hours.

Many procedures in DNA technology, e.g. DNA sequencing and profiling, require substantial amounts of DNA yet, very often, only small amounts are obtainable (e.g. DNA from a crime scene or from an extinct organism). **PCR** (**polymerase chain reaction**) is a technique for reproducing large quantities of DNA in the laboratory from an original sample. For this reason, it is often called **DNA amplification**. The technique is outlined below for a single cycle of replication. Subsequent cycles replicate DNA at an exponential rate, so PCR can produce billions of copies of DNA in only a few hours.

A single cycle of PCR



Repeat cycle of heating and cooling until enough copies of the target DNA have been produced

Loading tray
Prepared samples in tiny PCR tubes are placed in the loading tray and the lid is closed.

Temperature control
Inside the machine are heating and refrigeration mechanisms to rapidly change the temperature.

Dispensing pipette
Pipettes with disposable tips are used to dispense DNA samples into the PCR tubes.



RA

Thermal Cycler

Amplification of DNA can be carried out with simple-to-use machines called thermal cyclers. Once a DNA sample has been prepared, in just a few hours the amount of DNA can be increased billions of times. Thermal cyclers are in common use in the biology departments of universities, as well as other kinds of research and analytical laboratories. The one pictured on the left is typical of this modern piece of equipment.

DNA quantitation
The amount of DNA in a sample can be determined by placing a known volume in this quantitation machine. For many genetic engineering processes, a minimum amount of DNA is required.

Controls
The control panel allows a number of different PCR programmes to be stored in the machine's memory. Carrying out a PCR run usually just involves starting one of the stored programmes.

1. Explain the purpose of PCR: _____

_____

_____

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2. Describe how the **polymerase chain reaction** works: _____

3. Describe three situations where only very small DNA samples may be available for sampling and PCR could be used:

   (a) _____

   (b) _____

   (c) _____

4. After only two cycles of replication, four copies of the double-stranded DNA exist. Calculate how much a DNA sample will have increased after:

   (a) 10 cycles: _____ (b) 25 cycles: _____

5. The risk of contamination in the preparation for PCR is considerable.

   (a) Describe the effect of having a single molecule of unwanted DNA in the sample prior to PCR:

   (b) Describe two possible sources of DNA contamination in preparing a PCR sample:

   Source 1: _____

   Source 2: _____

   (c) Describe two precautions that could be taken to reduce the risk of DNA contamination:

   Precaution 1: _____

   Precaution 2: _____

6. Describe two other genetic engineering/genetic manipulation procedures that require PCR amplification of DNA:

   (a) _____

   (b) _____

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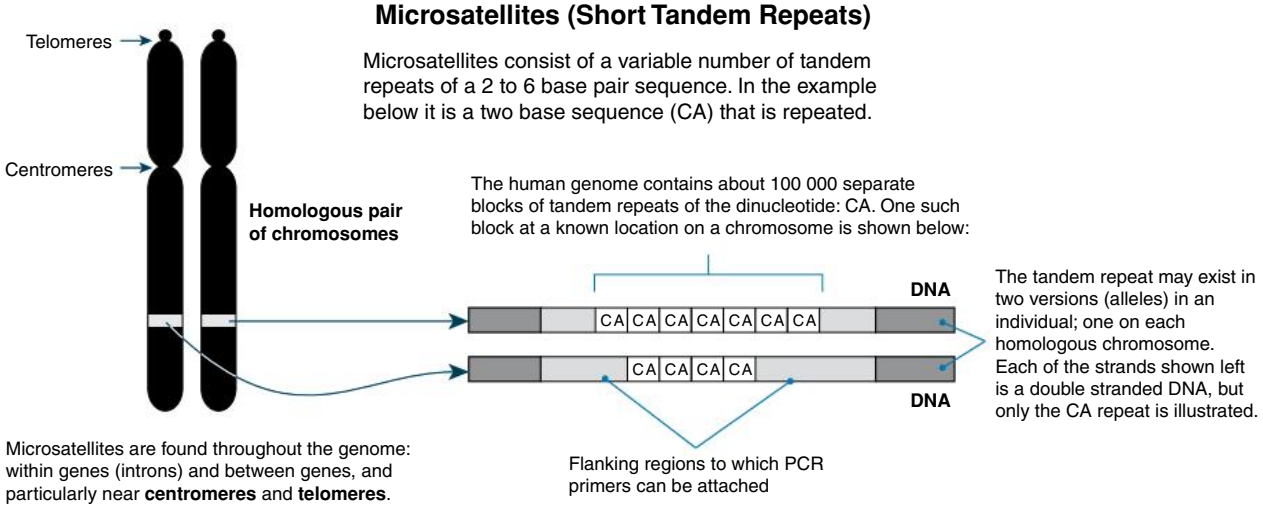
149 DNA Profiling Using PCR

Key Idea: Short units of DNA that repeat a different number of times in different people can be used to produce individual genetic profiles.

In chromosomes, some of the DNA contains simple, repetitive sequences. These non-coding nucleotide sequences repeat over and over again and are found scattered throughout the genome. Some repeating sequences, called **microsatellites** or **short tandem repeats** (STRs), are very short (2-6 base pairs) and can repeat up to 100 times. The human genome has many different microsatellites. Equivalent sequences in different people vary considerably in the numbers of the repeating unit. This phenomenon has been used to develop **DNA profiling**, which identifies the natural variations found in every person's DNA. Identifying these DNA differences is a useful tool for forensic investigations. The Victorian Institute of Forensic Medicine is one organisation offering forensic DNA testing in Australia. Tests target enough STR sites to guarantee that the odds of someone else sharing the same result are extremely unlikely, about one in a billion. DNA profiling has been used to help solve previously unsolved crimes and to assist in current or future investigations. DNA profiling can also be used to establish genetic relatedness (e.g. in paternity disputes or pedigree disputes), or when searching for a specific gene (e.g. screening for disease).

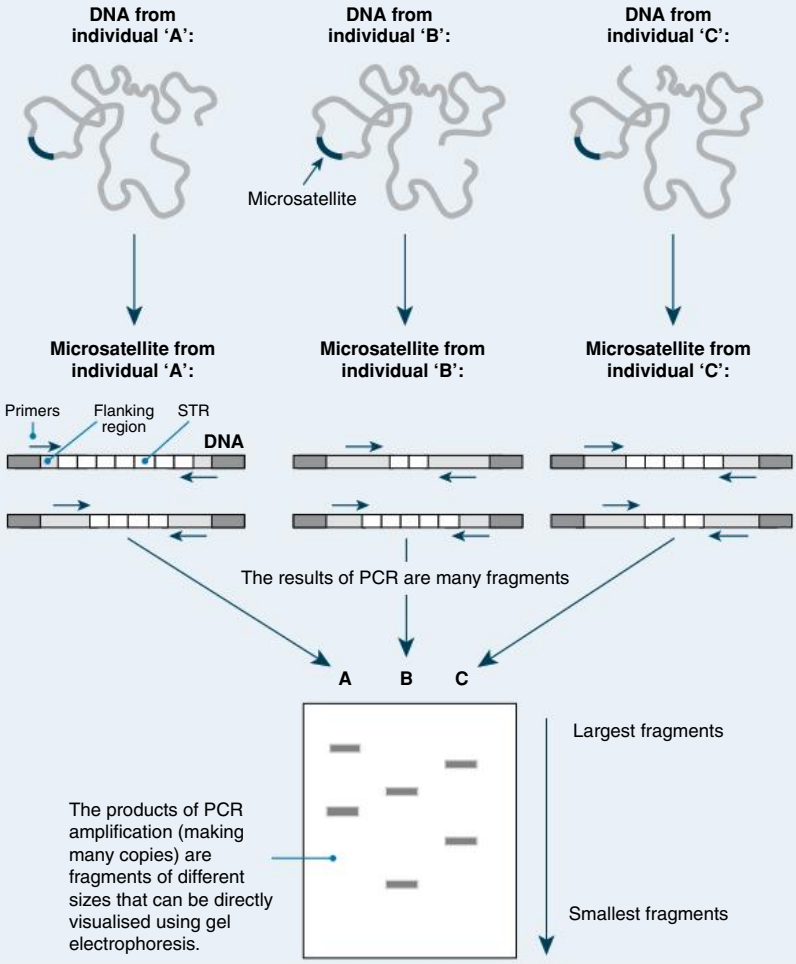
Microsatellites (Short Tandem Repeats)

Microsatellites consist of a variable number of tandem repeats of a 2 to 6 base pair sequence. In the example below it is a two base sequence (CA) that is repeated.



How short tandem repeats are used in DNA profiling

This diagram shows how three people can have quite different microsatellite arrangements at the same point (locus) in their DNA. Each will produce a different DNA profile using gel electrophoresis:



1 Extract DNA from sample

A sample collected from the tissue of a living or dead organism is treated with chemicals and enzymes to extract the DNA, which is separated and purified.

2 Amplify microsatellite using PCR

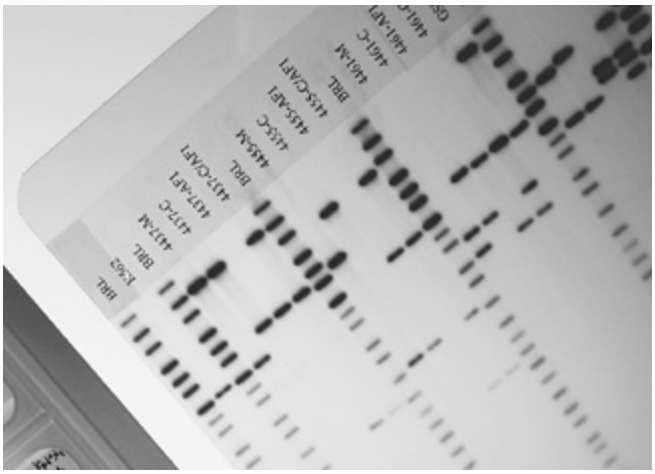
Specific primers (arrowed) that attach to the flanking regions (light grey) either side of the microsatellite are used to make large quantities of the micro-satellite and flanking regions sequence only (no other part of the DNA is amplified/replicated).

3 Visualise fragments on a gel

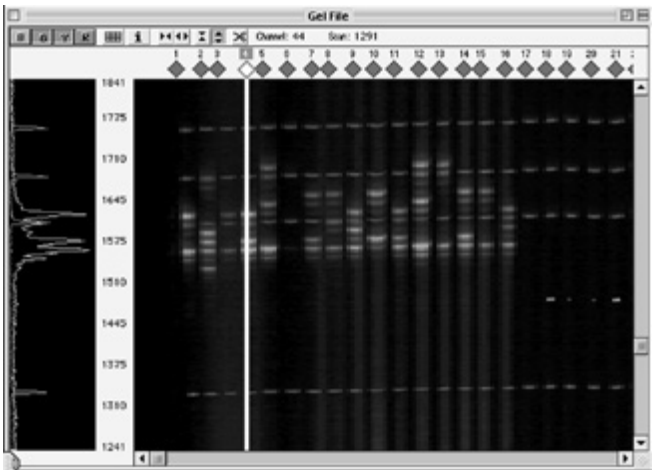
The fragments are separated by length, using **gel electrophoresis**. DNA, which is negatively charged, moves toward the positive terminal. The smaller fragments travel faster than larger ones.

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LINK 150 LINK 148 LINK 145 WEB 149 KNOW



The photo above shows a film output from a DNA profiling procedure. Those lanes with many regular bands are used for calibration; they contain DNA fragment sizes of known length. These calibration lanes can be used to determine the length of fragments in the unknown samples.



DNA profiling can be automated in the same way as DNA sequencing. Powerful computer software is able to display the results of many samples that are run at the same time. In the photo above, the sample in lane 4 has been selected and displays fragments of different length on the left of the screen.

1. Describe the properties of **short tandem repeats** that are important to the application of **DNA profiling** technology:

2. Explain the role of each of the following techniques in the process of DNA profiling:

   (a) Gel electrophoresis:

   (b) PCR:

3. Describe the three main steps in DNA profiling using PCR:

   (a)

   (b)

   (c)

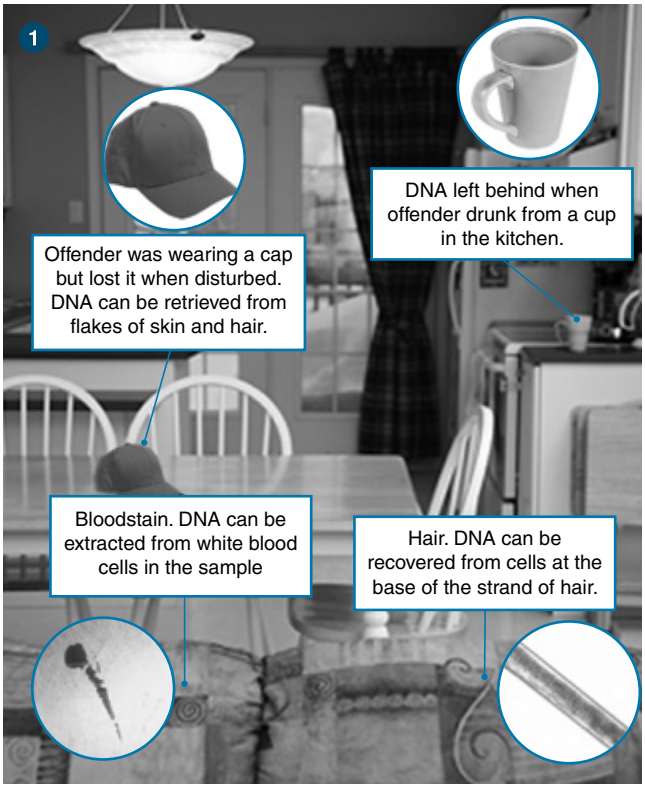
4. Explain why as many as 10 STR sites are used to gain a DNA profile for forensic evidence:

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150 Forensic Applications of DNA Profiling

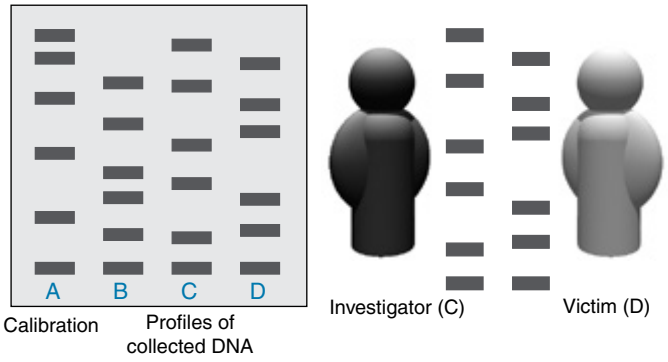
Key Idea: DNA profiling has many forensic applications, from identifying criminal offenders to saving endangered species.

The use of DNA as a tool for solving crimes such as homicide is well known, but it can also has several other applications. DNA evidence has been used to identify body parts, solve cases of industrial sabotage and contamination, for paternity testing, and even in identifying animal products illegally made from endangered species.

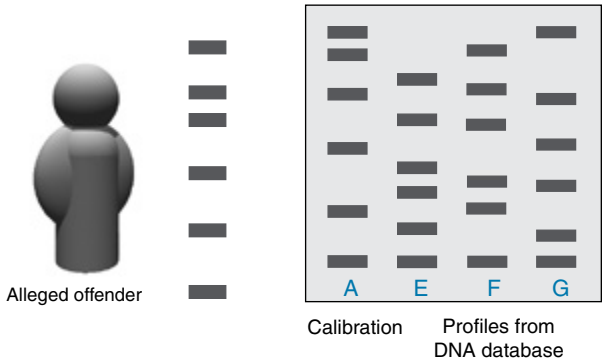


During the initial investigation, samples of material that may contain DNA are taken for analysis. At a crime scene, this may include blood and body fluids as well as samples of clothing or objects that the offender might have touched. Samples from the victim are also taken to eliminate them as a possible source of contamination.

2 DNA is isolated and profiles are made from all samples and compared to known DNA profiles such as that of the victim.



3 Unknown DNA samples are compared to DNA databases of convicted offenders and to the DNA of the alleged offender.



4 Although it does not make a complete case, DNA profiling, in conjunction with other evidence, is one of the most powerful tools in identifying offenders or unknown tissues.

1. Why are DNA profiles obtained for both the victim and investigator?

2. Use the evidence to decide if the alleged offender is innocent or guilty and explain your decision:

3. How could DNA profiling be used to refute official claims of the number of whales being captured and sold in fish markets?

Whale DNA: Tracking Illegal Slaughter



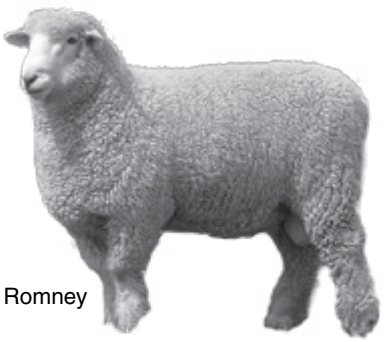
Under International Whaling Commission regulations, some species of whales can be captured for scientific research and their meat sold legally. Most, including humpback and blue whales, are fully protected and to capture or kill them for any purpose is illegal. Between 1999 and 2003 Scott Baker and associates from Oregon State University's Marine Mammal Institute investigated whale meat sold in markets in Japan and South Korea. Using DNA profiling techniques, they found around 10% of the samples tested were from fully protected whales including western gray whales and humpbacks. They also found that many more whales were being killed than were being officially reported.

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151 Finding the Connection

Key Idea: DNA technology has been used to identify the genetic relationships in and between many species.

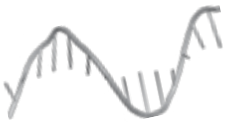
DNA technology has revolutionised the study of species relationships. Through genomic analyses, many so-called species that were once thought to be separate have been shown to be one. Conversely, taxa that were thought to be monospecific have now been shown to include more than one genetically distinct group. There are a number of different sheep breeds, each of which has been bred for a specific purpose (e.g. meat or wool production). As new sheep breeds are developed by selective breeding, older (traditional) breeds become less profitable to farm and are eventually replaced. These older breeds are becoming increasingly important because they carry traits that could be valuable but are now absent in more recent breeds. The relationships between the older and newer breeds are important as they show the development of breeds and help farmers and breeders to plan for the future development of their flocks.



Romney



UV tagged DNA primer 1



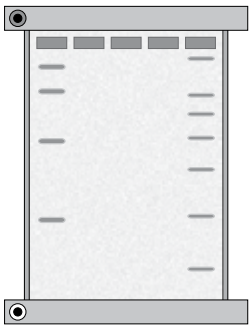
UV tagged DNA primer 2



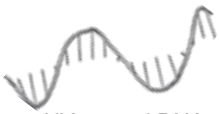
Merino

Connecting the Disconnected

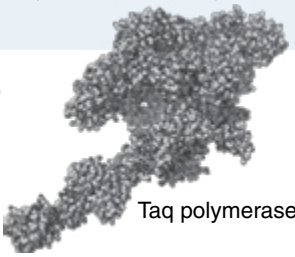
Your task is to use the items illustrated on this page to outline a technique to identify the genetic relationships between the following sheep breeds: Merino, Romney, Suffolk, Border Leicester, and Lincoln.



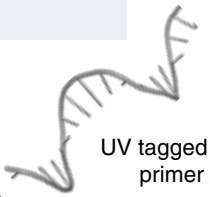
Electrophoresis gel



UV tagged DNA primer 3



Taq polymerase



UV tagged DNA primer 4



PCR machine

1. Identify the sections of the sheep DNA used to produce a DNA profile: _____

2. Explain the purpose of the DNA primers: _____

3. Describe briefly how identifiable pieces of DNA are isolated. _____

4. Explain the purpose of the electrophoresis gel in relation to this investigation: _____

5. Explain how the DNA profile produced can be used to identify relationships between the breeds of sheep:

LINK
TEST 149

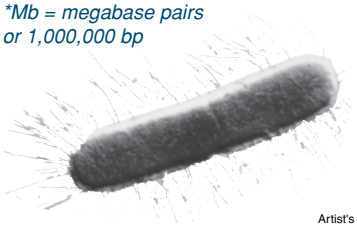
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152 Genome Projects

Key Idea: A genome is an organism's complete set of genetic material, including all of its genes. The genomes of many organisms have been sequenced, allowing genes to be compared. These can be searched for on gene databases.

The aim of most genome projects is to determine the DNA sequence of the organism's entire genome. Many different species have now had their genomes sequenced including the honeybee, nematode worm, African clawed frog, pufferfish, zebra fish, rice, cow, dog, and rat. Genome sizes and the number of genes per genome vary, and are not necessarily correlated with the size and structural complexity of the organism. Once completed, genome sequences are analysed by computer to identify genes. Gene sequences and details are often entered into online databases that can be searched by anyone wishing to find out information about a particular gene.

**Mb = megabase pairs or 1,000,000 bp*





Artist's impression





Bacterium
(*Escherichia coli*)

Genome size: 4.6 Mb*
Number of genes: 4403

E. coli has been used as a laboratory organism for over 70 years. Various strains of *E. coli* are responsible for several human diseases.

Yeast
(*Saccharomyces cerevisiae*)

Genome size: 13 Mb
Number of genes: 6000

The first eukaryotic genome to be completely sequenced. Yeast is used as a model organism to study human cancer.

Human
(*Homo sapiens*)

Genome size: 3000 Mb
Number of genes: < 22,500

The completion of the human genome has allowed advances in medical research, especially in cancer research.

Rice
(*Oryza sativa*)

Genome size: 466 Mb (indica) and 420 Mb (japonica)
Number of genes: 46,000

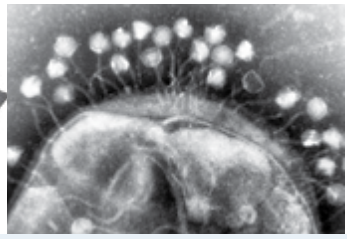
A food staple for much of the world's population. The importance of rice as a world food crop made sequencing it a high priority.







alpsdake PD



Graham Colm

Mouse
(*Mus musculus*)

Genome size: 2500 Mb
Number of genes: 30,000

New drugs destined for human use are often tested on mice because more than 90% of their proteins show similarities to human proteins.

Fruit fly
(*Drosophila melanogaster*)

Genome size: 150 Mb
Number of genes: 14,000

Drosophila has been used extensively for genetic studies for many years. About 50% of all fly proteins show similarities to mammalian proteins.

Japanese canopy plant
(*Paris japonica*)

Genome size: 149,000 Mb

This rare native Japanese plant has the largest genome sequenced so far (15% larger than any previous estimate for a eukaryote). Plants with very large genomes reproduce and grow slowly.

T2 phage

Genome size: 160,000 bp
Number of genes: Approx. 300

T2 phage is one of a group of related T-even phages that infect bacteria. Analysis of these phages indicates a small core genome with variations being the result of genetic transfers during evolution.

1. For each organism below, calculate how much smaller or larger the genome is than the human genome:

   (a) Japanese canopy plant: _____

   (b) *E. coli*: _____

   (c) T2 phage: _____

2. Plants with very large genome sizes are at higher risk of extinction. Can you suggest why? _____

   _____

3. Why would geneticists want to sequence the genomes of plants such as wheat, rice, and maize?

   _____

   _____

   _____

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153 Automated DNA Sequencing

Key Idea: The automation of DNA sequencing has helped to analyse the DNA sequences of many species.

The process of determining the DNA sequence of an organism can be automated using gel electrophoresis machines. Automation has made sequencing faster and less costly and thus has made large-scale genome sequencing projects possible. Automated sequencing analyses the light signals from nucleotides labelled with fluorescent dyes. Computer analysis of the data is then used to assemble the sequence. The basic automation method is outlined below. The new so-called next-generation high throughput sequencers extend this process across millions of reactions in parallel and are capable of producing thousands to millions of sequences at the same time.



1. DNA sample arrives

Purified DNA samples may contain linear DNA or plasmids. The sample should contain about 1 x 10^{11} DNA molecules. The sample is checked to ensure that there is enough DNA present in the sample to work with.



2. Primer and reaction mix added

A **DNA primer** is added to the sample which provides a starting sequence for synthesis. Also added is the **sequencing reaction mix** containing the *polymerase enzyme* and free nucleotides, some which are labelled with dye.



3. Create dye-labelled fragments

A **PCR** machine creates fragments of DNA complementary to the original template DNA. Each fragment is tagged with a fluorescent dye-labelled nucleotide. Running for 25 cycles, it creates 25x10^{11} single-stranded DNA molecules.

All photos are RA (unless indicated otherwise)



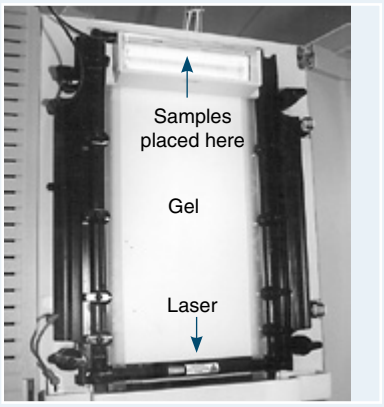
4. Centrifuge to create DNA pellet

The sample is chemically precipitated and centrifuged to settle the DNA fragments as a solid pellet at the bottom of the tube. Unused nucleotides, still in the liquid, are discarded.



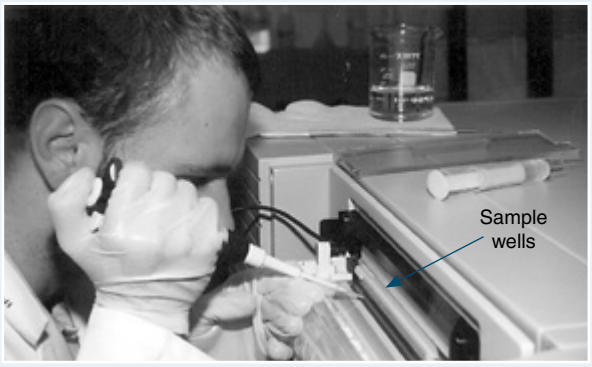
5. DNA pellet washed, buffer added

The pellet is washed with ethanol, dried, and a gel loading buffer is added. All that remains now is single stranded DNA with one dye-labelled nucleotide at the end of each molecule.



6. Acrylamide gel is loaded

The DNA sequencer is prepared by placing the gel (sandwiched between two sheets of glass) into position. A 36 channel 'comb' for receiving the samples is placed at the top of the gel.



BOB

7. Loading DNA samples onto gel

Different samples can be placed in each of the 36 wells (funnel shaped receptacles) above the gel. A control DNA sample of known sequence is applied to the first lane of the sequencer. If there are problems with the control sequence then results for all other lanes are considered invalid.



8. Running the DNA sequencer

Powerful computer software controls the activity of the DNA sequencer. The gel is left to run for up to 10 hours. During this time an argon laser is constantly scanning across the bottom of the gel to detect the passing of dye-labelled nucleotides attached to DNA fragments.

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The gel is loaded following preparation of the samples and the gel (see steps 1-7 and box, right).

Comb with 36 lanes into which different samples can be placed.

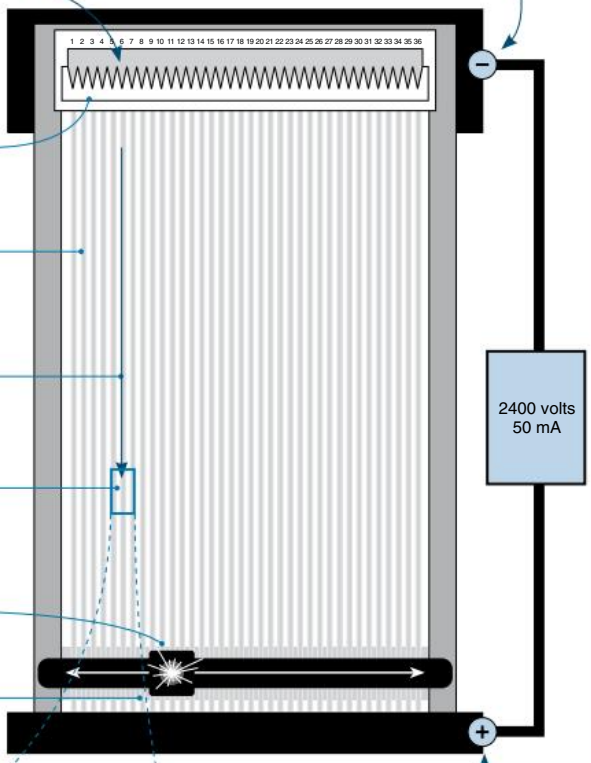
DNA fragments with dye-labelled nucleotides move down the gel over a period of 10 hours.

The smallest fragments move fastest down the gel and reach the argon laser first. Larger fragments arrive later.

DNA fragments separate into bands (see box below).

Argon laser excites fluorescent dye labels on nucleotides.

Lenses collect the emitted light and focus it into a spectrograph. An attached digital camera detects the light. See 'data collection' (below, right).



Creating the dye labelled fragments for gel electrophoresis is outlined in step 3. Key ingredients are:

(a) Original DNA template (the sample)

A C C G T A T G A T T C

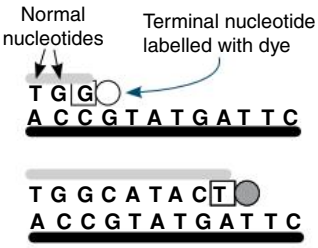
(b) Many normal unlabelled nucleotides:

A T G C

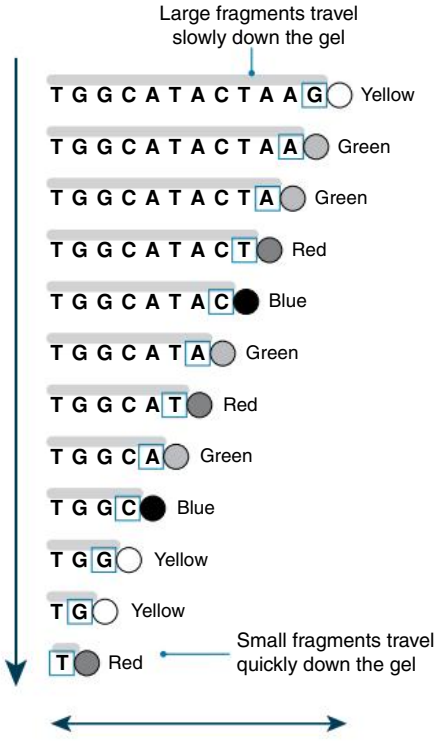
(c) Terminal nucleotides labelled with fluorescent dye (a different colour for each of the 4 bases). The structure of the nucleotides is altered so they act as terminators to stop further synthesis of the strand:



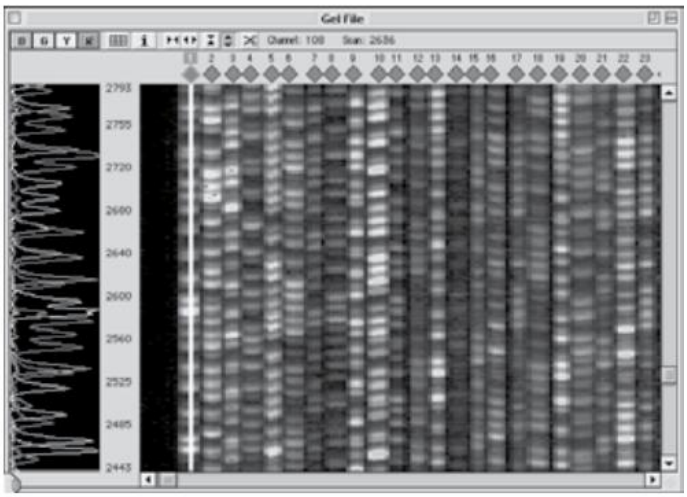
Two examples of synthesised DNA fragments are shown below. One is relatively short, the other is longer:



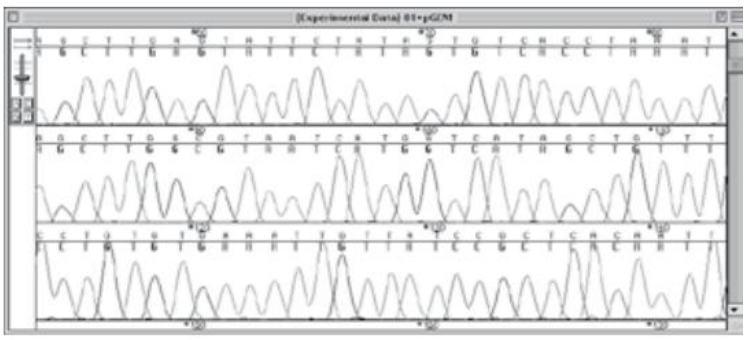
DNA fragments of different sizes are drawn down through the gel, separating into distinct bands of colour as they are illuminated by the laser:



Data collection: The data from the digital camera are collected by computer software. The first of 23 samples is highlighted below in lane 1 with base sequences appearing on the far left.



Data analysis: The data can be saved as a file for analysis by other computer software. Such software can provide a printout of the base sequence and perform comparisons with other DNA sequences (e.g. when looking for mutations).



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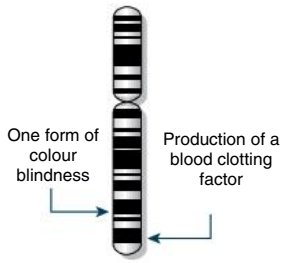
154 The Human Genome Project

Key Idea: The Human Genome Project (HGP) was a publicly funded global venture to determine the sequence of bases in the human genome and identify and map the genes.

The HGP was completed in 2003, ahead of schedule, although analysis of it continues. Other large scale sequencing projects have arisen as a result of the initiative to sequence the human genome. In 2002, for example, the International HapMap Project was started with the aim of describing the common patterns of human genetic variation. The HGP has provided an immense amount of information, but it is not the whole story. The next task is to find out what the identified genes do. The identification and study of the protein products of genes (**proteomics**) is an important development of the HGP and will give a better understanding of the functioning of the genome. There is also increasing research in the area of the human **epigenome**, which is the record of chemical changes to the DNA and histone proteins that do not involve changes in the DNA base sequence itself. The epigenome is important in regulating genome function. Understanding how it works is important to understanding disease processes.

Gene Mapping

This process involves determining the precise position of a gene on a chromosome. Once the position is known, it can be shown on a diagram.



X Chromosome

Equipment used for DNA Sequencing



Genesis Research and Development Corp. Auckland

Banks of PCR machines prepare the DNA for the sequencing gel stage. The DNA is amplified and chemically tagged (to make the DNA fluoresce and enable visualisation on a gel).



HGSI

Banks of DNA sequencing gels and powerful computers are used to determine the base order (the sequence) in DNA.

Count of Mapped Genes

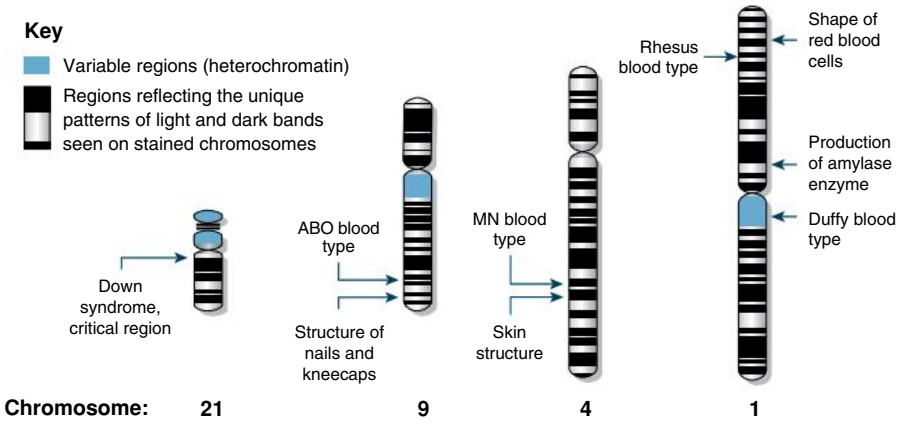
The aim of the HGP was to produce a continuous block of sequence information for each chromosome. Initially the sequence information was obtained to draft quality, with an error rate of 1 in 1000 bases. The **Gold Standard** sequence, with an error rate of <1 per 100,000 bases, was completed in October 2004. This table shows the length and number of mapped genes for each chromosome.

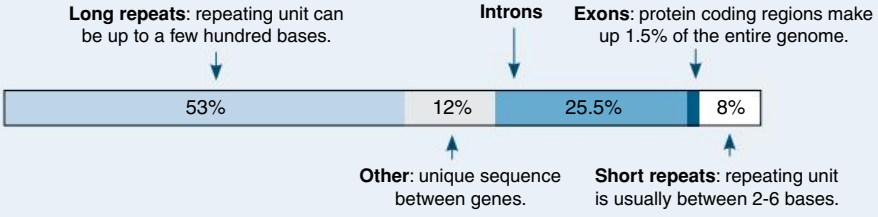
Chromosome	Length (Mb)	No. of Mapped Genes
1	263	1873
2	255	1113
3	214	965
4	203	614
5	194	782
6	183	1217
7	171	995
8	155	591
9	145	804
10	144	872
11	144	1162
12	143	894
13	114	290
14	109	1013
15	106	510
16	98	658
17	92	1034
18	85	302
19	67	1129
20	72	599
21	50	386
22	56	501
X	164	1021
Y	59	122
	Total:	**19,447**

Data to March 2008 from gdb.org (now offline)

Examples of Mapped Genes

The positions of an increasing number of genes have been mapped onto human chromosomes (see below). Sequence variations can cause or contribute to identifiable disorders. Note that chromosome 21 (the smallest human chromosome) has a relatively low gene density, while others are gene rich. This is possibly why trisomy 21 (Down syndrome) is one of the few viable human autosomal trisomies.





Key results of the HGP

- There are perhaps only 20,000-25,000 protein-coding genes in our human genome.
- It covers 99% of the gene containing parts of the genome and is 99.999% accurate.
- The new sequence correctly identifies almost all known genes (99.74%).
- Its accuracy and completeness allows systematic searches for causes of disease.

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Benefits and ethical issues arising from the Human Genome Project

Medical benefits

- Improved **diagnosis** of disease and predisposition to disease by genetic testing.
- Better identification of disease carriers, through genetic testing.
- Better **drugs** can be designed using knowledge of protein structure (from gene sequence information).
- Greater possibility of successfully using **gene therapy** to correct genetic disorders.

Non-medical benefits

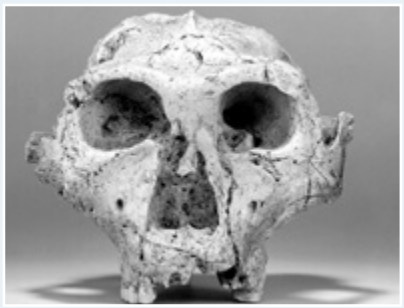
- Greater knowledge of **family relationships** through genetic testing, e.g. paternity testing in family courts.
- Advances **forensic science** through analysis of DNA at crime scenes.
- Better knowledge of the evolutionary relationships between humans and other organisms, which will help to develop more accurate classification systems.

Possible ethical issues

- Should third parties, e.g. health insurers, have rights to the results of genetic tests?
- Knowing your genetic predisposition for a disease is of no use is there is no treatment.
- Genetic tests are costly, and there is no easy answer as to who should pay for them.
- Genetic information is hereditary so knowledge of an individual's own genome has implications for members of their family.



Couples can already have a limited range of genetic tests to determine the risk of having offspring with some genetic disorders.



Comparative analysis of the sequence data for humans and their ancestors may provide clues about human evolution.



Legislation is needed to prevent discrimination on the basis of genetic profile, e.g. at work or for health insurance.

1. Briefly describe the objectives of the Human Genome Project (HGP): _____

2. Suggest a reason for developing a HapMap of the human genome: _____

3. Describe two possible **benefits** of Human Genome Project (HGP):

   (a) Medical: _____

   (b) Non-medical: _____

4. (a) What is **proteomics**? _____

   (b) Explain the significance of proteomics to the HGP and the ongoing benefits arising from it: _____

5. Suggest two possible points of view for one of the **ethical issues** described in the list above (top right):

   (a) _____

   (b) _____

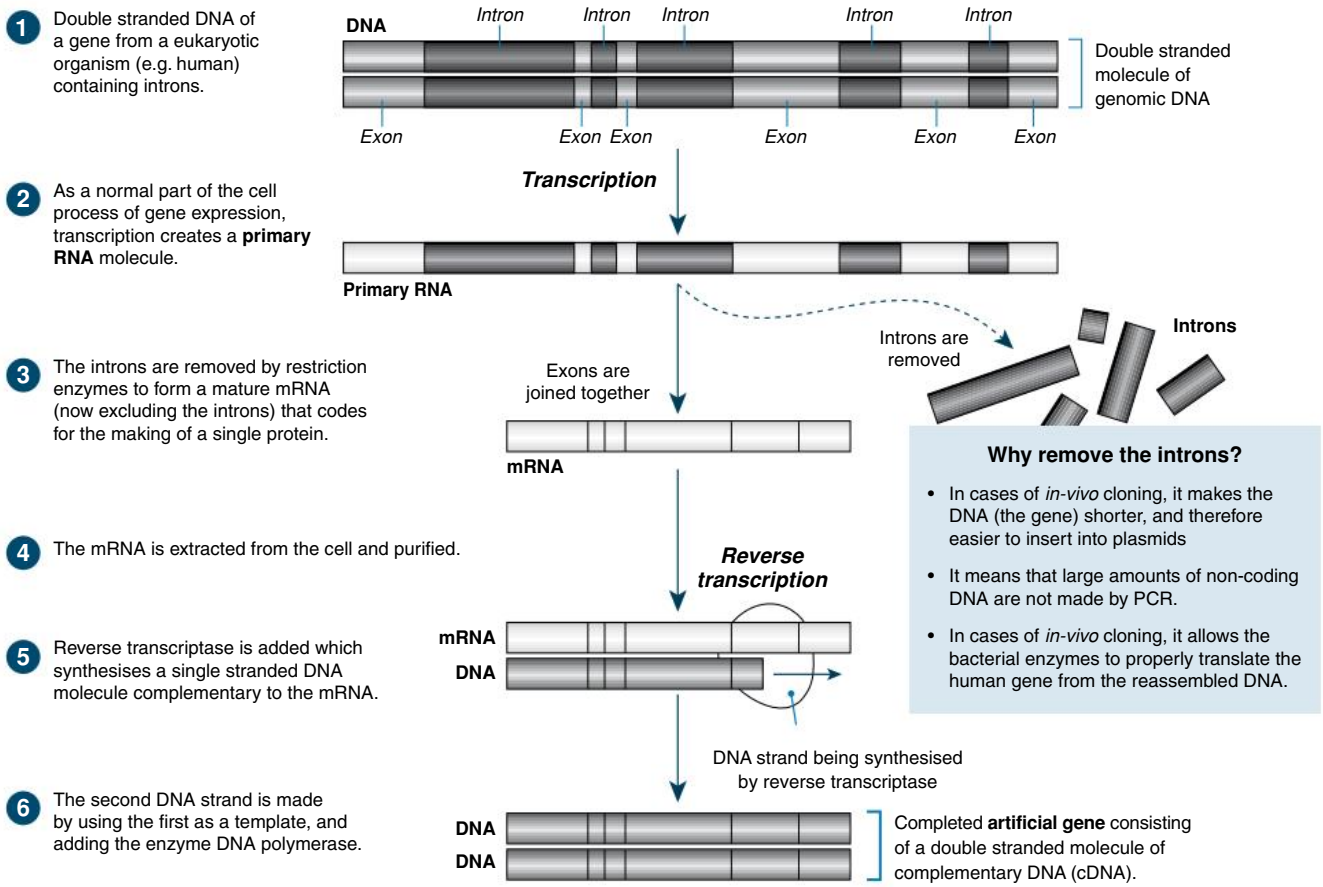
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155 Preparing a Gene for Cloning

Key Idea: Eukaryotic genes contain introns which must be removed before the gene can be inserted into a prokaryotic cell for *in vivo* cloning.

Not all the DNA in a eukaryotic gene is translated during protein synthesis. **Introns** are sequences of a gene's DNA that do not code for proteins. In eukaryotic cells, the introns are removed before mRNA from a transcribed gene is translated into a protein. The DNA sequences that are left behind are called **exons**, and the mRNA is called mature mRNA. Exons code for specific proteins. The presence of introns presents a problem when preparing a eukaryotic gene for insertion into a bacterial (prokaryotic) cell. Prokaryotic cells generally do not distinguish between introns and exons. The solution is to engineer a eukaryotic gene that can be transcribed and translated (expressed) by the prokaryote. This is achieved using the enzyme **reverse transcriptase**, which copies the mature mRNA (containing exons only) to produce a complementary strand of DNA. This task is important in both *in vivo* gene cloning and *ex vivo* gene cloning (by PCR) because it creates a gene ready for amplification.

Preparing a Gene for Cloning



1. What is the role of restriction enzymes in preparing a clone? _____

2. (a) Why are introns removed before cloning a gene? _____

   (b) What is the role of **reverse transcriptase** in this process? _____

3. What is the normal role of reverse transcriptase? _____

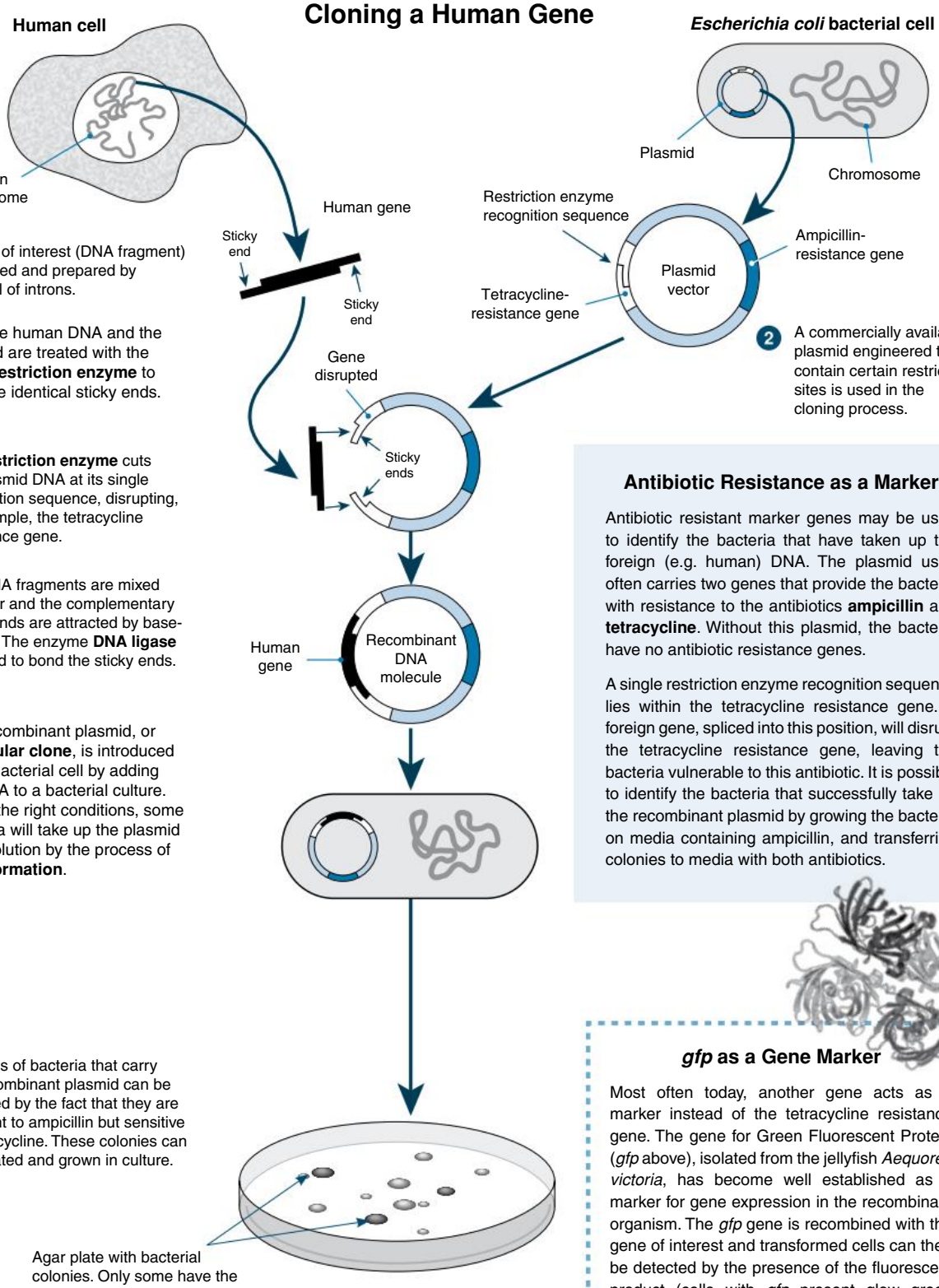
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156 *In Vivo* Gene Cloning

Key Idea: *In vivo* cloning describes the insertion of a gene into an organism and using the replication machinery of that organism to multiply the gene or produce its protein product.

Recombinant DNA techniques (restriction digestion and ligation) are used to insert a gene of interest into the DNA of a vector (e.g. plasmid or viral DNA). This produces a recombinant DNA molecule called a **molecular clone** that can transmit the gene of interest to another organism. To be useful, all vectors must be able to replicate inside their host organism, they must have one or more sites at which a restriction enzyme can cut, and they must have some kind of **genetic marker** that allows them to be identified. Bacterial plasmids are commonly used vectors because they are easy to manipulate, their restriction sites are well known, and they are readily taken up by cells in culture. Once the molecular clone has been taken up by bacterial cells, and those cells are identified, the gene can be replicated (cloned) many times as the bacteria grow and divide in culture.

Cloning a Human Gene



1. A gene of interest (DNA fragment) is isolated and prepared by removal of introns.
2. A commercially available plasmid engineered to contain certain restriction sites is used in the cloning process.
3. Both the human DNA and the plasmid are treated with the same **restriction enzyme** to produce identical sticky ends.
4. The **restriction enzyme** cuts the plasmid DNA at its single recognition sequence, disrupting, for example, the tetracycline resistance gene.
5. The DNA fragments are mixed together and the complementary sticky ends are attracted by base-pairing. The enzyme **DNA ligase** is added to bond the sticky ends.
6. The recombinant plasmid, or **molecular clone**, is introduced into a bacterial cell by adding the DNA to a bacterial culture. Under the right conditions, some bacteria will take up the plasmid from solution by the process of **transformation**.
7. Colonies of bacteria that carry the recombinant plasmid can be identified by the fact that they are resistant to ampicillin but sensitive to tetracycline. These colonies can be isolated and grown in culture.

Antibiotic Resistance as a Marker

Antibiotic resistant marker genes may be used to identify the bacteria that have taken up the foreign (e.g. human) DNA. The plasmid used often carries two genes that provide the bacteria with resistance to the antibiotics **ampicillin** and **tetracycline**. Without this plasmid, the bacteria have no antibiotic resistance genes.

A single restriction enzyme recognition sequence lies within the tetracycline resistance gene. A foreign gene, spliced into this position, will disrupt the tetracycline resistance gene, leaving the bacteria vulnerable to this antibiotic. It is possible to identify the bacteria that successfully take up the recombinant plasmid by growing the bacteria on media containing ampicillin, and transferring colonies to media with both antibiotics.



gfp as a Gene Marker

Most often today, another gene acts as a marker instead of the tetracycline resistance gene. The gene for Green Fluorescent Protein (*gfp* above), isolated from the jellyfish *Aequorea victoria*, has become well established as a marker for gene expression in the recombinant organism. The *gfp* gene is recombined with the gene of interest and transformed cells can then be detected by the presence of the fluorescent product (cells with *gfp* present glow green under fluorescent light).

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1. Explain why it might be desirable to use *in vivo* methods to clone genes rather than PCR:

2. Explain when it may not be desirable to use bacteria to clone genes:

3. Explain how a human gene is removed from a chromosome and placed into a plasmid.

4. A bacterial plasmid replicates at the same rate as the bacteria. If a bacteria containing a recombinant plasmid replicates and divides once every thirty minutes, calculate the number of plasmid copies there will be after twenty four hours:

5. When cloning a gene using **plasmid vectors**, the bacterial colonies containing the recombinant plasmids are mixed up with colonies that have none. All the colonies look identical, but some have taken up the plasmids with the human gene, and some have not. Explain how the colonies with the recombinant plasmids are identified:

6. Explain why the *gfp* marker is a more desirable gene marker than genes for antibiotic resistance:

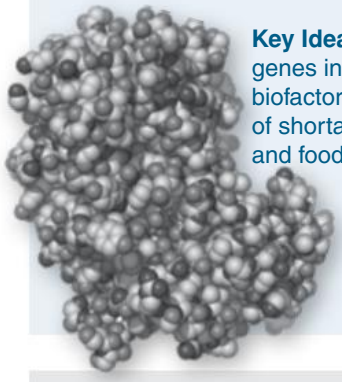
7. Bacteriophages are viruses that infect bacteria:

   (a) What feature of bacteriophages make them useful for genetic engineering?

   (b) How could a bacteriophage be used to clone a gene?

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157 Using Recombinant Bacteria



Key Idea: Inserting useful genes into bacteria to produce biofactories can solve the problem of shortages in the manufacturing and food industries.

The Issue

- **Chymosin** (also known as **rennin**) is an enzyme that digests milk proteins. It is the active ingredient in rennet, a substance used by cheesemakers to clot milk into curds.
- Traditionally rennin is extracted from "chyme", i.e. the stomach secretions of suckling calves (hence its name of chymosin).
- By the 1960s, a shortage of chymosin was limiting the volume of cheese produced.
- Enzymes from fungi were used as an alternative but were unsuitable because they caused variations in the cheese flavour.

Concept 1

Enzymes are proteins made up of amino acids. The amino acid sequence of chymosin can be determined and the mRNA coding sequence for its translation identified.

Concept 2

Reverse transcriptase can be used to synthesise a DNA strand from the mRNA. This process produces DNA without the introns, which cannot be processed by bacteria.

Concept 3

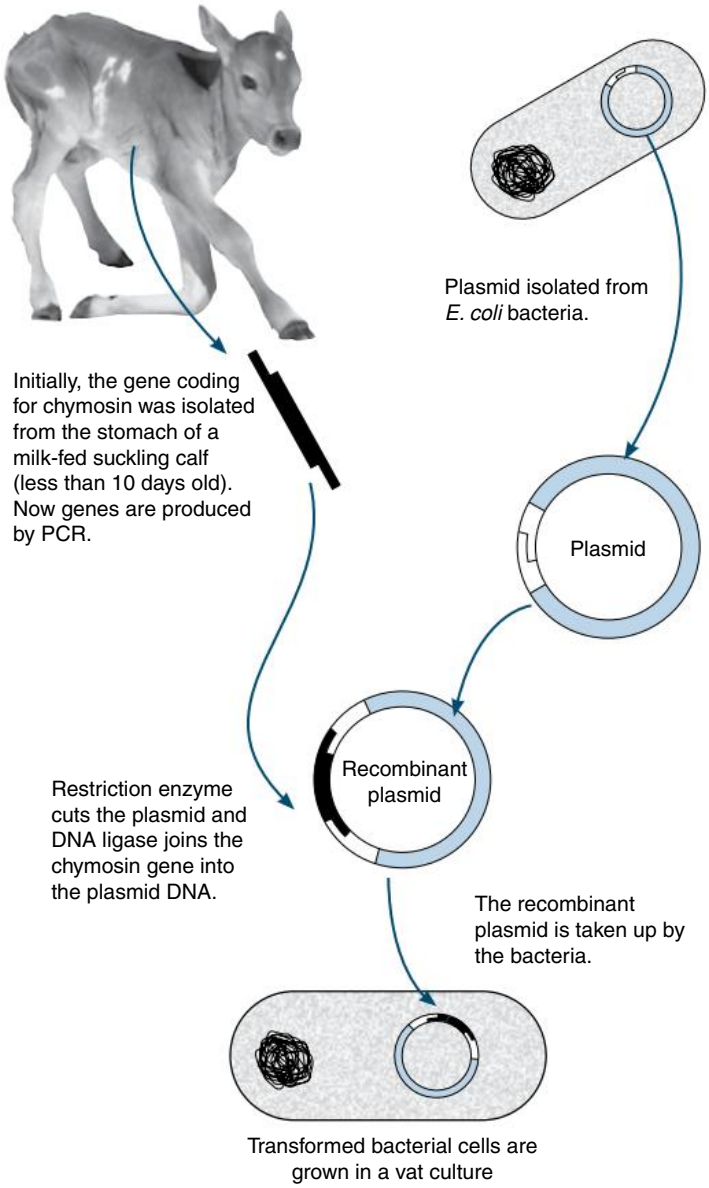
DNA can be cut at specific sites using **restriction enzymes** and rejoined using **DNA ligase.** New genes can be inserted into self-replicating bacterial **plasmids**.

Concept 4

Under certain conditions, bacteria are able to lose or take up plasmids from their environment. Bacteria are readily grown in vat cultures at little expense.

Concept 5

The protein in made by the bacteria in large quantities.



Techniques

The amino acid sequence of chymosin is first determined and the RNA codons for each amino acid identified.

mRNA matching the identified sequence is isolated from the stomach of young calves. **Reverse transcriptase** is used to transcribe mRNA into DNA. The DNA sequence can also be made synthetically once the sequence is determined.

The DNA is amplified using PCR.

Plasmids from *E. coli* bacteria are isolated and cut using **restriction enzymes.** The DNA sequence for chymosin is inserted using **DNA ligase**.

Plasmids are returned to *E. coli* by placing the bacteria under conditions that induce them to take up plasmids.

Outcomes

The transformed bacteria are grown in vat culture. Chymosin is produced by *E. coli* in packets within the cell that are separated during the processing and refining stage.

Recombinant chymosin entered the marketplace in 1990. It established a significant market share because cheesemakers found it to be cost effective, of high quality, and in consistent supply. Most cheese is now produced using recombinant chymosin such as CHY-MAX.

Further Applications

A large amount of processing is required to extract chymosin from *E.coli.* There are now a number of alternative bacteria and fungi that have been engineered to produce the enzyme. Most chymosin is now produced using the fungi ***Aspergillus niger*** and ***Kluyveromyces lactis***. Both are produced in a similar way as that described for *E. coli.*

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Enzymes from GMOs are widely used in the baking industry. Maltogenic alpha amylase from *Bacillus subtilis* bacteria is used as an anti-staling agent to prolong shelf life. Hemicellulases from *B. subtilis* and xylanase from the fungus *Aspergillus oryzae* are used for improvement of dough, crumb structure, and volume during the baking process.



Romain Behar

Lipase from *Aspergillus oryzae* is used in processing of palm oil to produce low cost cocoa butter substitutes (above), which have a similar 'mouth feel' to cocoa butter.



Dual Freq

Acetolactate decarboxylase from *B. subtilis* is an enzyme used in the brewing industry. It reduces maturation time of the beer by by-passing a rate-limiting step.

1. Describe the main use of chymosin: _____

_____

2. What was the traditional source of chymosin? _____

_____

3. Summarise the key concepts that led to the development of the technique for producing chymosin:

   (a) Concept 1: _____

   (b) Concept 2: _____

   (c) Concept 3: _____

   (d) Concept 4: _____

   (e) Concept 5: _____

4. Discuss how the gene for chymosin was isolated and how the technique could be applied to isolating other genes:

_____

_____

_____

_____

_____

_____

5. Describe three advantages of using chymosin produced by GE bacteria over chymosin from traditional sources:

   (a) _____

   _____

   (b) _____

   _____

   (c) _____

   _____

6. Explain why the fungus *Aspergillus niger* is now more commonly used to produce chymosin instead of *E. coli*:

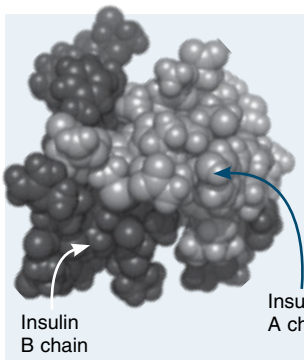
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158 Insulin Production



Key Idea: By using microorganisms to make human insulin, problematic issues of cost, allergic reactions, and ethics have been addressed.

The Issue

- **Type I diabetes mellitus** is a metabolic disease caused by a lack of **insulin**. Around 25 people in every 100,000 suffer from type I diabetes.
- It is treatable only with injections of insulin.
- In the past, insulin was taken from the pancreases of cows and pigs and purified for human use. The method was expensive and some patients had severe allergic reactions to the foreign insulin or its contaminants.

Concept 1

DNA can be cut at specific sites using **restriction enzymes** and joined together using **DNA ligase**. Genes can be inserted into self-replicating bacterial **plasmids** at the point where the cuts are made.

Concept 2

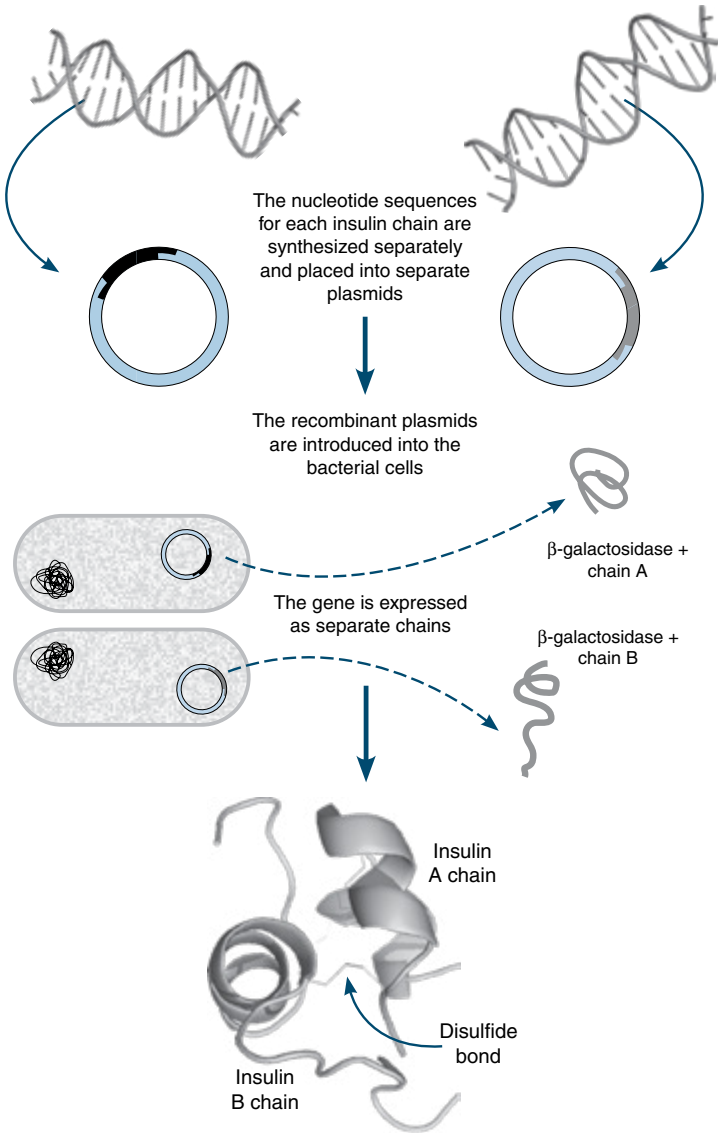
Plasmids are small, circular pieces of DNA found in some bacteria. They usually carry genes useful to the bacterium. *E. coli* plasmids can carry promoters required for the transcription of genes.

Concept 3

Under certain conditions, Bacteria are able to lose or pick up plasmids from their environment. Bacteria can be readily grown in vat cultures at little expense.

Concept 4

The DNA sequences coding for the production of the two polypeptide chains (A and B) that form human insulin can be isolated from the human genome.



Techniques

The **gene** is **chemically synthesised** as two nucleotide sequences, one for the **insulin A chain** and one for the **insulin B chain**. The two sequences are small enough to be inserted into a plasmid.

Plasmids are extracted from *Escherichia coli*. The gene for the bacterial enzyme **β-galactosidase** is located on the plasmid. To make the bacteria produce insulin, the insulin gene must be linked to the **β-galactosidase** gene, which carries a promoter for transcription.

Restriction enzymes are used to cut plasmids at the appropriate site and the A and B insulin sequences are inserted. The sequences are joined with the plasmid DNA using **DNA ligase**.

The **recombinant plasmids** are inserted back into the bacteria by placing them together in a culture that favours plasmid uptake by bacteria.

The bacteria are then grown and multiplied in vats under carefully controlled growth conditions.

Outcomes

The product consists partly of β-galactosidase, joined with either the A or B chain of insulin. The chains are extracted, purified, and mixed together. The A and B insulin chains connect via **disulfide cross linkages** to form the functional insulin protein. The insulin can then be made ready for injection in various formulations.

Further Applications

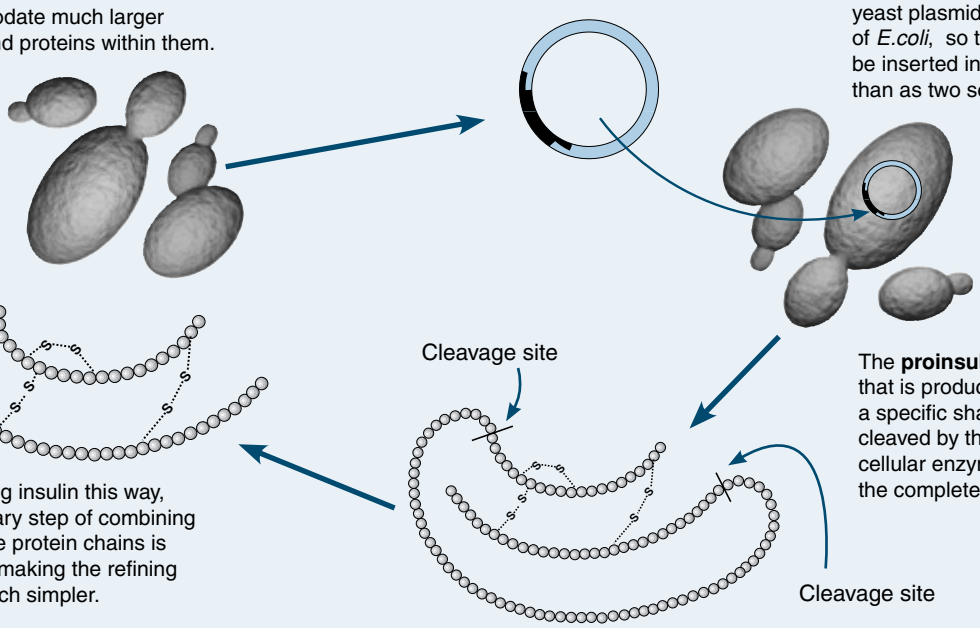
The techniques used to produce human insulin from genetically modified bacteria can be applied to a range of human proteins and hormones. Proteins currently being produced include human growth hormone, interferon, and factor VIII.

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Insulin Production in *Saccharomyces*

Yeast cells are **eukaryotic** and hence are much larger than bacterial cells. This enables them to accommodate much larger plasmids and proteins within them.



The gene for human insulin is inserted into a plasmid. The yeast plasmid is larger than that of *E.coli*, so the entire gene can be inserted in one piece rather than as two separate pieces.

The **proinsulin** protein that is produced folds into a specific shape and is cleaved by the yeast's own cellular enzymes, producing the completed insulin chain.

Cleavage site

Cleavage site

By producing insulin this way, the secondary step of combining the separate protein chains is eliminated, making the refining process much simpler.

1. Describe the three major problems associated with the traditional method of obtaining insulin to treat diabetes:

   (a) _____

   (b) _____

   (c) _____

2. Explain the reasoning behind using *E. coli* to produce insulin and the benefits that GM technology has brought to diabetics:

   _____

   _____

   _____

   _____

3. Explain why, when using *E. coli*, the insulin gene is synthesised as two separate A and B chain nucleotide sequences:

   _____

   _____

4. Why are the synthetic nucleotide sequences (‘genes’) 'tied' to the β-galactosidase gene? _____

   _____

   _____

5. Yeast (*Saccharomyces cerevisiae*) is also used in the production of human insulin. Discuss the differences in the production of insulin using yeast and *E. coli* with respect to:

   (a) Insertion of the gene into the plasmid: _____

   _____

   _____

   (b) Secretion and purification of the protein product: _____

   _____

   _____

   _____

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159 Genetically Modified Food Plants



Key Idea: The use of recombinant DNA to build a new metabolic pathway has greatly increased the nutritional value of a rice.

The Issue

- **Beta-carotene** (β-carotene) is a precursor to **vitamin A** which is involved in many functions including vision, immunity, fetal development, and skin health.
- Vitamin A deficiency is common in developing countries where up to 500,000 children suffer from night blindness, and death rates due to infections are high due to a lowered immune response.
- Providing enough food containing useful quantities of β-carotene is difficult and expensive in many countries.

Concept 1

Rice is a staple food in many developing countries. It is grown in large quantities and is available to most of the population, but it lacks many of the essential nutrients required by the human body for healthy development. It is low in β-carotene.

Concept 2

Rice plants produce β-carotene but not in the edible rice **endosperm**. Engineering a new biosynthetic pathway would allow β-carotene to be produced in the endosperm. Genes expressing enzymes for carotene synthesis can be inserted into the rice genome.

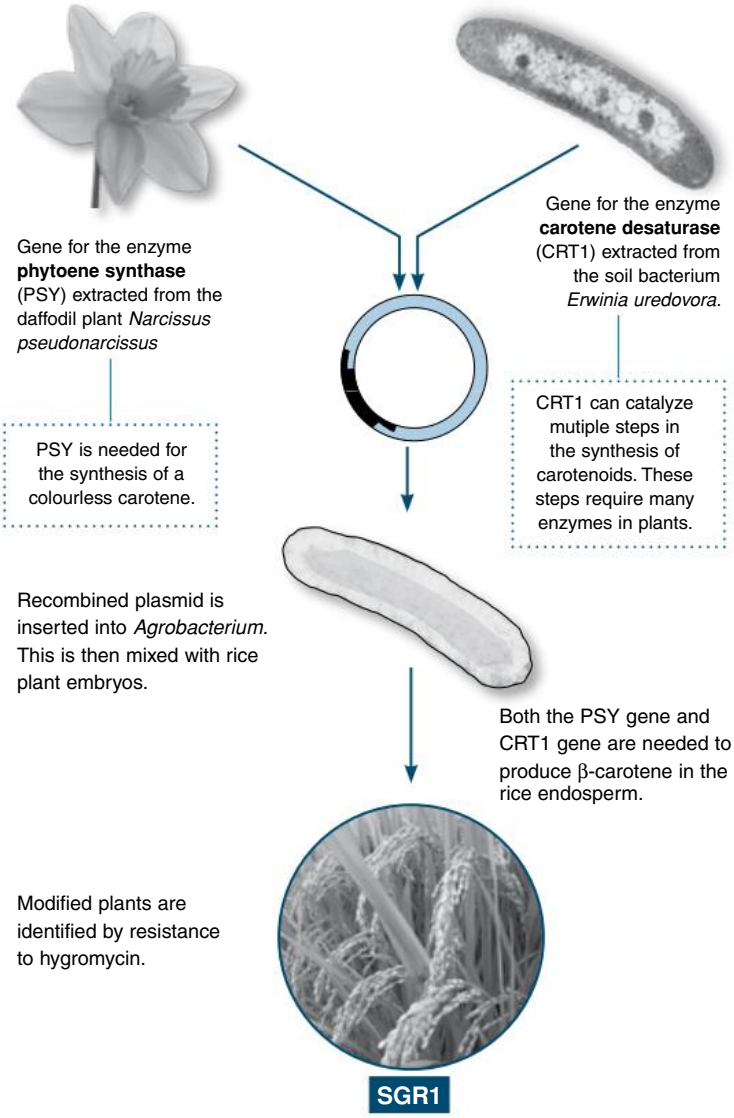
Concept 3

The enzyme **carotene desaturase (CRT1)** in the soil bacterium *Erwinia uredovora,* catalyses multiple steps in carotenoid biosynthesis. **Phytoene synthase (PSY)** overexpresses a colourless carotene in the daffodil plant *Narcissus pseudonarcissus.*

Concept 4

DNA can be inserted into an organism's genome using a suitable **vector**. ***Agrobacterium tumefaciens*** is a tumour-forming bacterial plant pathogen that is commonly used to insert novel DNA into plants.

The Development of Golden Rice



Techniques

The **PSY** gene from daffodils and the **CRT1** gene from *Erwinia uredovora* are sequenced.

DNA sequences are synthesised into packages containing the CRT1 or PSY gene, terminator sequences, and **endosperm specific promoters** (these ensure expression of the gene only in the edible portion of the rice).

The ***Ti* plasmid** from *Agrobacterium* is modified using restriction enzymes and DNA ligase to delete the tumour-forming gene and insert the synthesized DNA packages. A gene for resistance to the antibiotic **hygromycin** is also inserted so that transformed plants can be identified later. The parts of the *Ti* plasmid required for plant transformation are retained.

Modified *Ti* plasmid is inserted into the bacterium.

Agrobacterium is incubated with rice plant embryo. Transformed embryos are identified by their resistance to hygromycin.

Outcomes

The rice produced had endosperm with a distinctive yellow colour. Under greenhouse conditions golden rice (**SGR1**) contained 1.6 µg per g of carotenoids. Levels up to five times higher were produced in the field, probably due to improved growing conditions.

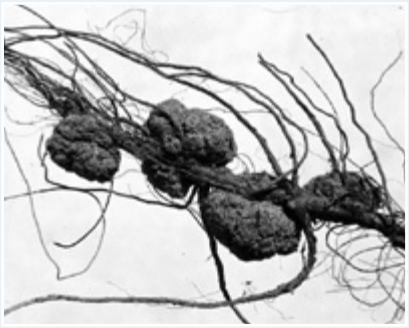
Further Applications

Further research on the action of the PSY gene identified more efficient methods for the production of β-carotene. The second generation of golden rice now contains up to 37 µg per g of carotenoids. Golden rice was the first instance where a complete biosynthetic pathway was engineered. The procedures could be applied to other food plants to increase their nutrient levels.

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All photos USDA



The ability of *Agrobacterium* to transfer genes to plants is exploited for crop improvement. The tumour-inducing *Ti* plasmid is modified to delete the tumour-forming gene and insert a gene coding for a desirable trait. The parts of the *Ti* plasmid required for plant transformation are retained.



Soybeans are one of the many food crops that have been genetically modified for broad spectrum herbicide resistance. The first GM soybeans were planted in the US in 1996. By 2007, nearly 60% of the global soybean crop was genetically modified; the highest of any other crop plant.



GM cotton was produced by inserting the gene for the BT toxin into its genome. The bacterium *Bacillus thuringiensis* naturally produces BT toxin, which is harmful to a range of insects, including the larvae that eat cotton. The BT gene causes cotton to produce this insecticide in its tissues.

1. Describe the basic methodology used to create golden rice: _____

2. Explain how scientists ensured β-carotene was produced in the endosperm: _____

3. What property of *Agrobacterium tumefaciens* makes it an ideal vector for introducing new genes into plants?

4. (a) How could this new variety of rice reduce disease in developing countries? _____

   (b) Absorption of vitamin A requires sufficient dietary fat. Explain how this could be problematic for the targeted use of golden rice in developing countries:

5. As well as increasing nutrient content as in golden rice, other traits of crop plants are also desirable. For each of the following traits, suggest features that could be desirable in terms of increasing yield:

   (a) Grain size or number: _____

   (b) Maturation rate: _____

   (c) Pest resistance: _____

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160 Food for the Masses

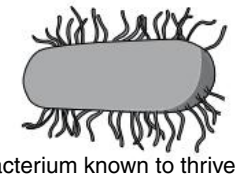
Key Idea: Genetic engineering has the potential to solve many of the world's food shortage problems by producing crops with greater yields than those currently grown.

Currently 1/6 of the world's population are **undernourished**. If trends continue, 1.5 billion people will be at risk of starvation by 2050 and, by 2100 (if global warming is taken into account), nearly half the world's population could be threatened with food shortages. The solution to the problem of food production is complicated. Most of the Earth's arable land has already been developed and currently uses 37% of the Earth's land area, leaving little room to grow more crops or farm more animals. Development of new fast growing and high yield crops appears to be part of the solution, but many crops can only be grown under a narrow range of conditions or are susceptible to disease. Moreover, the farming and irrigation of some areas is difficult, costly, and can be environmentally damaging. **Genetic modification** of plants may help to solve some of these problems by producing plants that will require less intensive culture or that will grow in areas previously considered not arable.

Useful Organisms



Fungus that is able to survive dry conditions using two enzymes WA-UT1 and Ter-UT2 to facilitate water uptake.

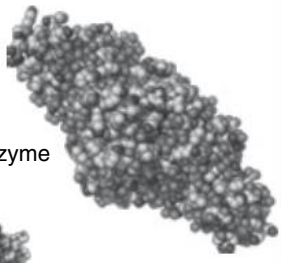


Bacterium known to thrive in dry conditions using a single enzyme DRI-X1 to catalyse multiple reactions.

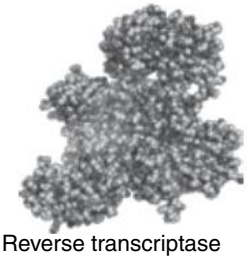


Plant identified for modification

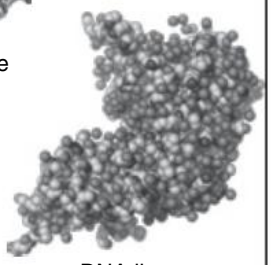
Enzymes



Restriction enzyme



Reverse transcriptase



DNA ligase

Engineering Your Solution

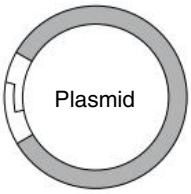
A solution to the possible future food crisis is to genetically engineer food crops so that they can maximise their growth under adverse conditions. Standard selective breeding techniques could be used to do this, but in some plants this may not be possible or feasible and it may require more time than is available. A selection of genetic tools and organisms with useful characteristics are described. **Your task** is to use the items shown to devise a technique to successfully create a plant that could be successfully farmed in semi-desert environments such as sub-Saharan Africa. The following page will take you through the procedure. Not all the items will need to be used.



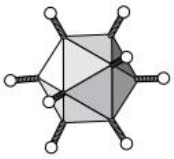
Petri dish



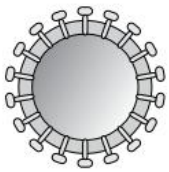
Incubator



Equipment



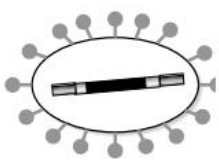
Adenovirus



Retrovirus



Agrobacterium



Liposome

Possible Vectors

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1. Identify the organism you would chose as a 'donor' of drought survival genes and explain your choice:

2. Describe a process to identify and isolate the required gene(s) and identify the tools to be used:

3. Identify a vector for the transfer of the isolated gene(s) into the crop plant and explain your decision:

4. Explain how the isolated gene(s) would be integrated into the vector's genome:

5. (a) Explain how the vector will transform the identified plant:

   (b) Identify the stage of development at which the plant would most easily be transformed. Explain your choice:

6. Explain how the transformed plants could be identified:

7. Explain how a large number of plants can be grown from the few samples that have taken up the new DNA:



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161 The Ethics of GMO Technology

Key Idea: There are many potential benefits and risks in using genetically modified organisms.

Genetically modified organisms (GMOs) have many potential benefits, but their use raises a number of biological and ethical concerns. Some of these include risk to human health, animal welfare issues, and environmental safety. Currently a matter of concern to consumers is the adequacy of government regulations for the labelling of food products with GMO content. In some countries GM products must be clearly labelled, while other countries have no requirements for GM labelling. This can take away consumer choice about the types of products they buy. The use of GM may also have trade implications for countries exporting and importing GMO produce.

Some important points about GMOs

1. The modified DNA is in every cell of the GMO.
2. The mRNA is only expressed in specific tissues.
3. The foreign protein is only expressed in particular tissues but it may circulate in the blood or lymph or be secreted (e.g. milk).
4. In animals, the transgene is only likely to be transmitted from parent to offspring. However, viral vectors may enable accidental transfer of the transgene between unrelated animals.
5. In plants, transmission of the transgene in GMOs is possible by pollen, cuttings, and seeds (even between species).
6. If we eat the animal or plant proper, we will also be eating DNA. The DNA will remain 'intact' if raw, but "degraded" if cooked.
7. Non-transgenic food products may be processed using genetically modified bacteria or yeast, and cells containing their DNA may be in the food product.
8. A transgenic product (e.g. a protein, polypeptide or a carbohydrate) may be in the GMO, but not in the portions sold to the consumer.

Potential effects of GMOs on the world

1. Increase in food production.
2. Decrease in use of pesticides, herbicides and animal remedies.
3. Improvement in the health of the human population and the medicines used to achieve it.
4. Possible development of transgenic products which may be harmful to some (e.g. new proteins causing allergies).
5. May have little real economic benefit to farmers (and the consumer) when increased production (as little as 10%) is weighed against cost, capital, and competition.
6. Possible (uncontrollable) spread of transgenes into other species: plants, indigenous species, animals, and humans.
7. Concerns that the release of GMOs into the environment may be irreversible.
8. Economic sanctions resulting from a consumer backlash against GMO foods and products.
9. Animal welfare and ethical issues: GM animals may suffer poor health and reduced life span.
10. GMOs may cause the emergence of pest, insect, or microbial resistance to traditional control methods.
11. May create a monopoly and dependence of developing countries on companies who are seeking to control the world's commercial seed supply.



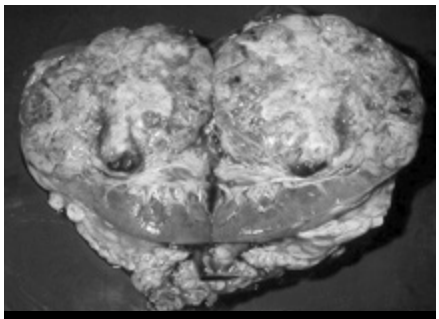
© Greenpeace

GMO protestors are arrested

Issue: The accidental release of GMOs into the environment.

Problem: Recombinant DNA may be taken up by non-target organisms. These then may have the potential to become pests or cause disease.

Solution: Rigorous controls on the production and release of GMOs. GMOs could have specific genes deleted so that their growth requirements are met only in controlled environments.



CDC

Cancerous kidney

Issue: A new gene or genes may disrupt normal gene function.

Problem: Gene disruption may trigger cancer. Successful expression of the desired gene is frequently very low.

Solution: A combination of genetic engineering, cloning, and genetic screening so that only those cells that have been successfully transformed are used to produce organisms.



© Greenpeace

Protest against GMOs in the environment

Issue: Targeted use of transgenic organisms in the environment.

Problem: Once their desired function, e.g. environmental clean-up, is completed, they may be undesirable invaders in the ecosystem.

Solution: GMOs can be engineered to contain "suicide genes" or metabolic deficiencies so that they do not survive for long in the new environment after completion of their task.

1. Suggest why genetically modified (GM) plants are thought to pose a greater environmental threat than GM animals:

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2. Describe an advantage and a problem with the use of genetically engineered herbicide resistant crop plants:

   (a) Advantage: _____

   _____

   (b) Problem: _____

   _____

3. Describe an advantage and a problem with using tropical crops genetically engineered to grow in cold regions:

   (a) Advantage: _____

   _____

   (b) Problem: _____

   _____

4. Describe an advantage and a problem with using crops that are genetically engineered to grow in marginal habitats (for example, in very saline or poorly aerated soils):

   (a) Advantage: _____

   _____

   (b) Problem: _____

   _____

5. Describe two uses of transgenic animals within the livestock industry:

   (a) _____

   (b) _____

6. Some years ago, Britain banned the import of a GM, pest resistant corn variety containing marker genes for ampicillin antibiotic resistance. Suggest why the use of antibiotic-resistance genes as markers is no longer common practice:

   _____

   _____

7. Many agricultural applications of DNA technology make use of transgenic bacteria which infect plants and express a foreign gene. Explain one advantage of each of the following applications of genetic engineering to crop biology:

   (a) Development of nitrogen-fixing *Rhizobium* bacteria that can colonise non-legumes such as corn and wheat:

   _____

   (b) Addition of transgenic *Pseudomonas fluorescens* bacteria into seeds (bacterium produces a pathogen-killing toxin):

   _____

8. Some of the public's fears and concerns about genetically modified food stem from moral or religious convictions, while others have a biological basis and are related to the potential biological threat posed by GMOs.
   (a) Conduct a class discussion or debate to identify these fears and concerns, and list them below:

   _____

   _____

   _____

   _____

   (b) Identify which of those you have listed above pose a real biological threat: _____

   _____

   _____

   _____



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162 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its definition, as identified by its preceding letter code.

amplification .....

annealing .....

blunt end .....

DNA ligation .....

DNA polymerase .....

DNA profiling .....

gel electrophoresis .....

gene technology .....

GMO .....

marker gene .....

microsatellite .....

molecular clone .....

PCR .....

plasmid .....

primer .....

recognition site .....

recombinant DNA .....

restriction enzyme .....

sticky end .....

vector .....

A An organism or artificial vehicle that is capable of transferring a DNA sequence to another organism.

B The pairing (by hydrogen bonding) of complementary single-stranded nucleic acids to form a double-stranded polynucleotide. The term is applied to making recombinant DNA, to the binding of a DNA probe, or to the binding of a primer to a DNA strand during PCR.

C The process of locating regions of a DNA sequence that are variable between individuals in order to distinguish between individuals.

D A short length of DNA used to identify the starting sequence for PCR so that polymerase enzymes can begin amplification.

E An enzyme that is able to cut a length of DNA at a specific sequence or site.

F One of multiple copies of an isolated defined sequence of (often recombinant) DNA.

G A gene, with an identifiable effect, used to determine if a piece of DNA has been successfully inserted into the host organism.

H A reaction that is used to amplify fragments of DNA using cycles of heating and cooling (abbreviation).

I A small circular piece of DNA commonly found in bacteria.

J The process of producing more copies of a length of DNA, normally using PCR.

K DNA that has had a new sequence added so that the original sequence has been changed.

L The repairing or attaching of fragmented DNA by ligase enzymes.

M A short (normally two base pairs) piece of DNA that repeats a variable number of times between people and so can be used to distinguish between individuals.

N A type of cut in a length of DNA caused by a restriction enzyme that results in both strands of DNA being the same length.

O An enzyme that is able to replicate DNA and commonly used in PCR to amplify a length of DNA.

P An organism that has had part of its DNA sequence altered either by the removal or insertion of a piece of DNA.

Q A process that is used to separate different lengths of DNA by placing them in a gel matrix placed in a buffered solution through which an electric current is passed.

R The manipulation of DNA and gene sequences in order to modify the characteristics of organisms.

S The site or sequence of DNA at which a restriction enzyme attaches and cuts the DNA.

T A cut in a length of DNA by a restriction enzyme that results in two strands of DNA being different lengths with one strand overhanging the other.

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QLD As required
VCE Unit 4 (1)
WA ATAR Unit 3
SA Theme O

Regulation of Gene Expression

Key terms

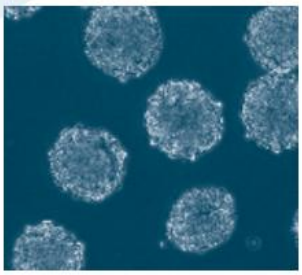
adult stem cells
apoptosis
cancer
cellular differentiation
embryonic stem cells
gene expression
homeotic genes
Hox genes
inducible genes
morphogenesis
morphogens
mutation
p53
operon
oncogenes
regulatory sequence
stem cells
transcription factors
tumour suppressor genes

Timing and coordination of development

Learning aims and skills

Activity number

- ☐ 1 Explain how changes in gene expression result in the cellular differentiation of stem cells during development of the embryo as well as later in life. — 163
- ☐ 2 Recall the role of apoptosis in normal cell differentiation and morphogenesis. Describe the consequences of excessive or insufficient apoptosis. — 163 168
- ☐ 3 Explain the role of homeotic genes, including the *Hox* genes, in developmental patterning in organisms. Suggest why these gene sequences are highly conserved across animal phyla. — 164
- ☐ 4 Explain how embryonic induction during development results in developmental events occurring at the correct time in the correct sequence. — 165
- ☐ 5 Describe an example of a mutation that results in errors in development or gene regulation. Examples could include the genetic hearing loss or cancer. — 166 168







Gene regulation

Learning aims and skills

Activity number

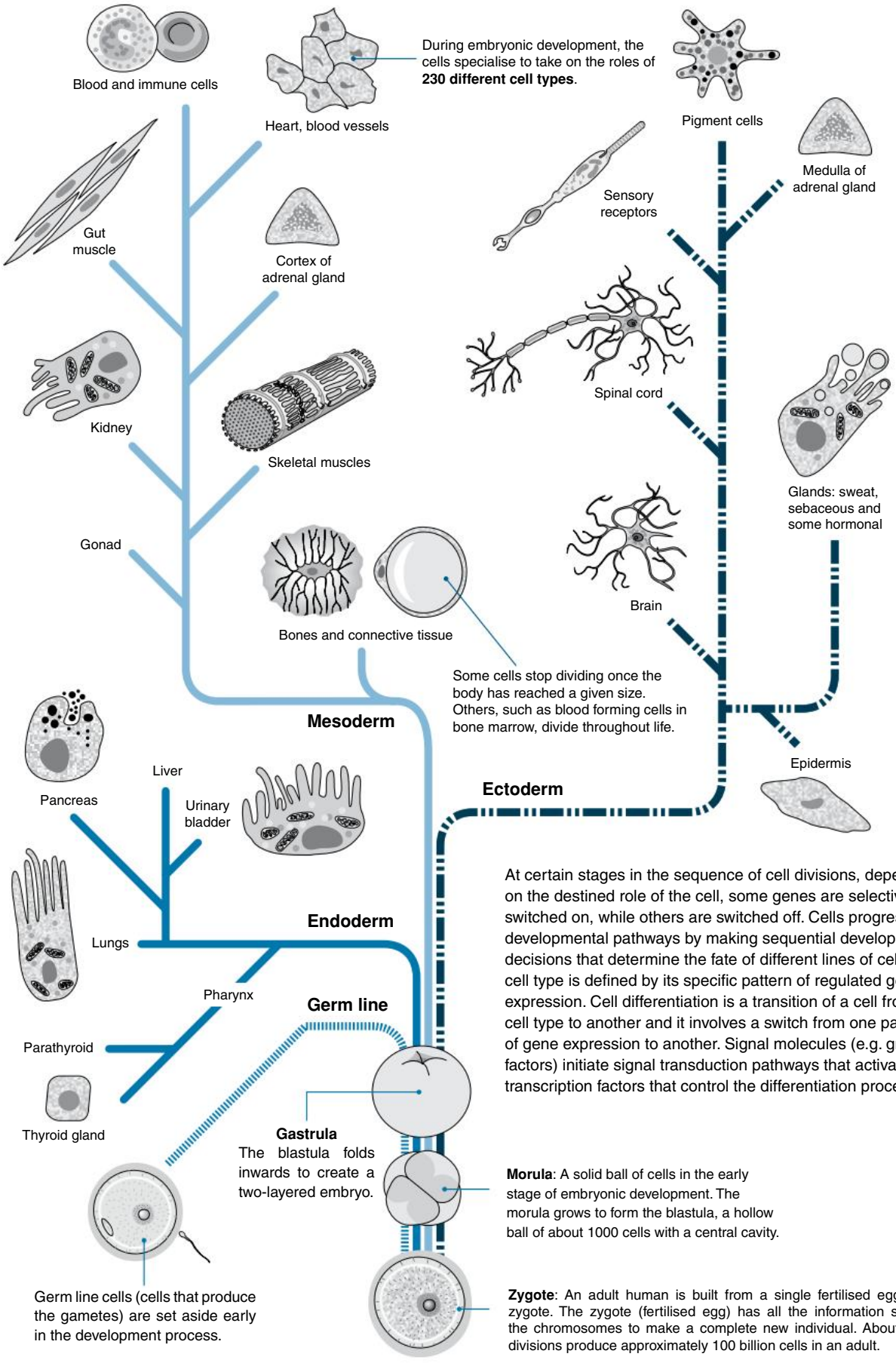
- ☐ 6 Describe how gene expression in bacteria is regulated by operons. Recognise an operon as a cluster of genes under the control of a single promoter. Identify parts of the operon and their roles in regulating transcription. Describe the role of the regulator gene in producing the repressor molecule and explain what the repressor molecule does. — 167
- ☐ 7 Explain the regulation of gene expression in eukaryotes, including the role of transcription factors. Recall that differences in gene regulation account for some of the phenotypic differences between genomically similar organisms. — 167
- ☐ 8 Explain the role of tumour suppressor genes and proto-oncogenes in regulating cell division. Explain how carcinogens can upset these regulatory mechanisms and trigger tumour formation (cancer). — 168



163 Cellular Differentiation

Key Idea: A zygote divides and produces all the cell types in the body by cellular differentiation.

A zygote begins development by dividing into a small ball of a few dozen identical cells called **embryonic stem cells**. These cells start to take different developmental paths to become specialised cells, such as nerve cells, and can no longer produce another cell type. **Differentiation** is the end result of this developmental pathway.



At certain stages in the sequence of cell divisions, depending on the destined role of the cell, some genes are selectively switched on, while others are switched off. Cells progress along developmental pathways by making sequential developmental decisions that determine the fate of different lines of cells. Each cell type is defined by its specific pattern of regulated gene expression. Cell differentiation is a transition of a cell from one cell type to another and it involves a switch from one pattern of gene expression to another. Signal molecules (e.g. growth factors) initiate signal transduction pathways that activate transcription factors that control the differentiation process.

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Development

Development is the process of progressive change through the lifetime of an organism. It involves growth (increase in size) and cell division (to generate the multicellular body). **Cellular differentiation** (the generation of specialised cells from more generalised ones) and **morphogenesis** (the creation of the shape and form of the body) are also part of development.



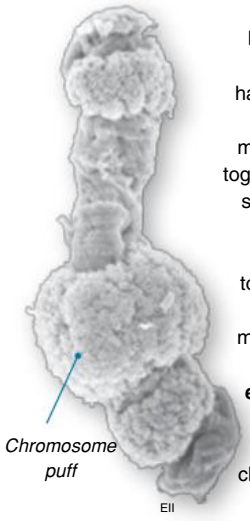
Mouse embryo 12.5 days

Matthias Zepper

Selective cell proliferation combined with selective apoptosis sculpts the tissues and structures in all vertebrates. In the mouse embryo, mesoderm forms between the toes giving the appearance of a webbed, paddle like structure (above left). As the embryo develops, this superfluous webbing is selectively destroyed by apoptosis so that each of the individual digits are visible (above right, arrowed).

Control Over Genes

Gene expression is the process by which a cell regulates the production of gene products (DNA or RNA) to meet the cell's requirements. Regulation is achieved by switching the transcription of specific genes on and off, starting or stopping the production of the gene product so that it is produced only when it is needed. Gene activity is regulated by cell type, cell function, chemical signals, and signals from the environment.



Many insect larvae have large **polytene chromosomes** (chromosomes which have undergone repeated rounds of DNA replication without cell division so that many sister chromatids remain synapsed together). They contain many copies of the same gene, so have a high level of gene expression. For example, *Chironomus* larvae produce large amounts of saliva to aid digestion of detritus. As they moult and grow, salivary gland development must keep pace with their increased food requirements. The moulting hormone, **ecdysone**, acts as the **signal** to turn on the gene regulating the development of the salivary glands. The regions of the chromosome with this gene puff out (left) when the genes are being transcribed.

1. Germ line cells diverge (become isolated) from other cells at a very early stage in embryonic development:

   (a) Explain what the **germ line** is: _____

   (b) Explain why it is necessary for the germ line to become separated at such an early stage of development:

2. Explain the genetic events that enable so many different cell types to arise from one unspecialised cell (the zygote):

3. Using an example, describe the role of signal molecules in activating gene expression at the appropriate time:

4. Explain briefly the importance of gene regulation in development: _____

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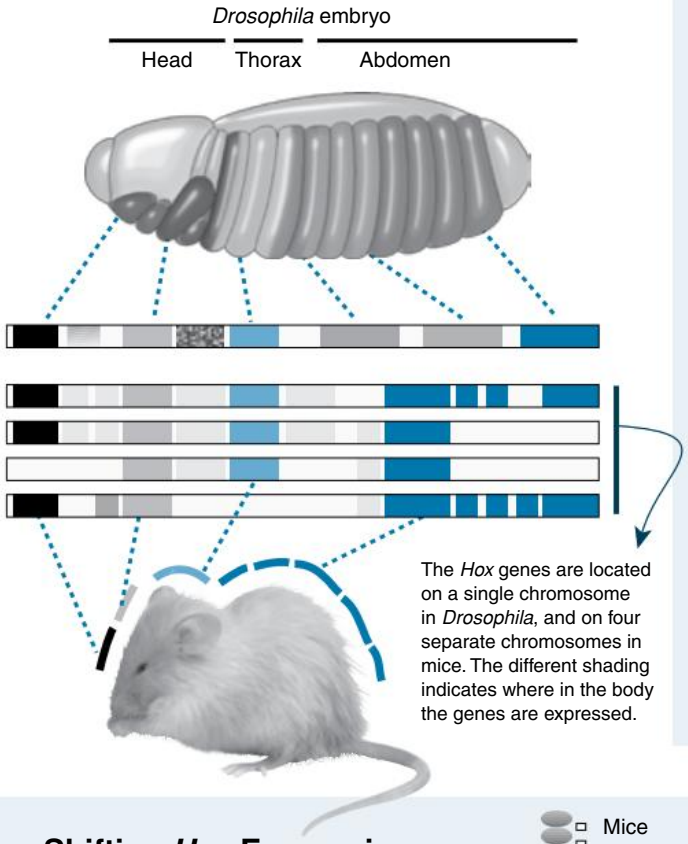
164 Homeotic Genes and Development

Key Idea: Small changes to the regulation of genes can lead to large scale changes in the development of organisms.

The **homeobox** is region of highly conserved gene sequences, which direct development by regulating gene expression. The ***Hox* genes** are a group of homeobox genes in animals. They are found in clusters and determine where body segments and limbs grow. Very different organisms share this same set of genes, but regulate them differently. This means that large changes in morphology or function are attributable to changes in gene regulation, rather than the evolution of new genes, and natural selection associated with gene switches plays a major role in evolution.

The Role of *Hox* Genes

Hox genes control the development of back and front parts of the body. The same genes (or homologous ones) are present in essentially all animals, including humans.



The *Hox* genes are located on a single chromosome in *Drosophila*, and on four separate chromosomes in mice. The different shading indicates where in the body the genes are expressed.

The Evolution of Novel Forms

Even very small changes (mutations) in the *Hox* genes can have a profound effect on morphology. Such changes to the genes controlling development have almost certainly been important in the evolution of novel structures and body plans. Four principles underlie the role of developmental genes in the evolution of novel forms:

- **Evolution works with what is already present**: New structures are modifications of pre-existing structures.
- **Multifunctionality** and **redundancy**: Functional redundancy in any part of a multifunctional structure allows for specialisation and division of labour through the development of two separate structures.

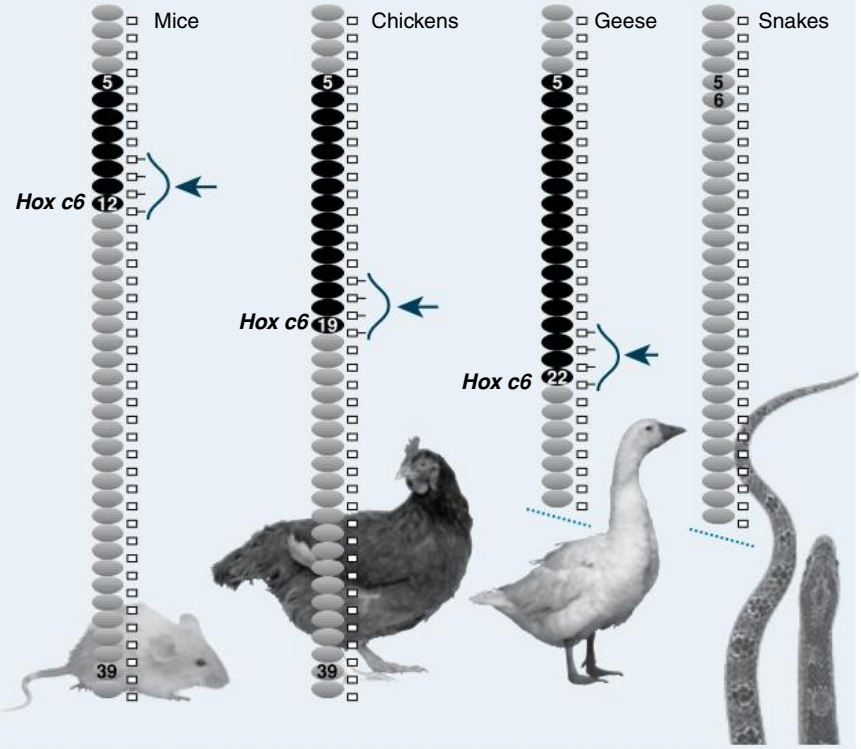
  Example: the diversity of appendages (including mouthparts) in arthropods.
- **Modularity**: Modular architecture in animals (arthropods, vertebrates) allows for the modification and specialization of individual body parts. Genetic switches allow changes in one part of a structure, independent of other parts.



Shifting *Hox* Expression

Huge diversity in morphology in organisms within and across phyla could have arisen through small changes in the genes controlling development.

Differences in neck length in vertebrates provide a good example of how changes in gene expression can bring about changes in morphology. Different vertebrate species have different numbers of neck vertebrae (denoted by the black ovals on the diagram). The boundary between neck and trunk vertebrae is marked by expression of the ***Hox c6* gene** (c6 denotes the sixth cervical or neck vertebra) in all cases, but the position varies in each animal relative to the overall body. The forelimb (arrow) arises at this boundary in all four-legged vertebrates. In snakes, the boundary is shifted forward to the base of the skull and no limbs develop. As a result of these differences in expression, mice have a short neck, geese a long neck, and snakes, no neck at all.

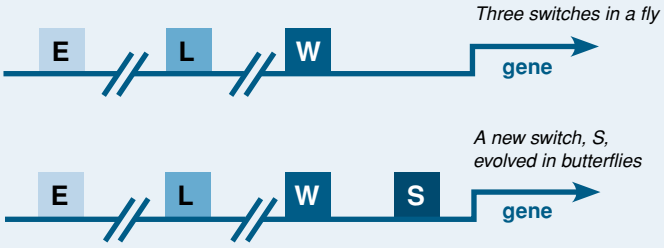


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Genetic Switches in Evolution

The *Hox* genes are just part of the collection of genes that make up the genetic tool kit for animal development. The genes in the tool kit act as switches, shaping development by affecting how other genes are turned on or off. The distribution of genes in the tool kit indicates that it is ancient and was in place before the evolution of most types of animals. Differences in form arise through changes in genetic switches. One example is the evolution of eyespots on the wings of butterflies:

- The ***Distal-less*** gene is one of the important **master body-building genes** in the genetic tool kit. Switches in the *Distal-less* gene control expression in the embryo (E), larval legs (L), and wing (W) in flies and butterflies, but butterflies have also evolved an extra switch (S) to control eyespot development.
- Once spots evolved, changes in *Distal-less* expression (through changes in the switch) produced more or fewer spots.



Changes in *Distal-less* regulation were probably achieved by changing specific sequences of the *Distal-less* gene eyespot switch. The result? Changes in eyespot size and number.

Same Gene, New Tricks



Stichophthalma camadeva | *Junonia coenia (buckeye)* | *Taenaris macrops*

- The action of a tool kit protein depends on context: where particular cells are located at the time when the gene is switched on.
- Changes in the DNA sequence of a genetic switch can change the zone of gene expression without disrupting the function of the tool kit protein itself.
- The spectacular **eyespots** on butterfly wings (arrowed above) represent different degrees of a basic pattern, from virtually all eyespot elements expressed (*Stichophthalma*) to very few (*Taenaris*).

1. Briefly describe the role of homeobox genes in development: _____

2. (a) What does it mean when the homeobox genes are said to be highly conserved? _____

   (b) What does this tell you about the evolution and the importance of the homeobox sequences? _____

3. Suggest why the *Hox* genes are found in clusters (i.e. grouped tightly together): _____

4. Using an example, discuss how changes in gene expression can bring about changes in morphology:

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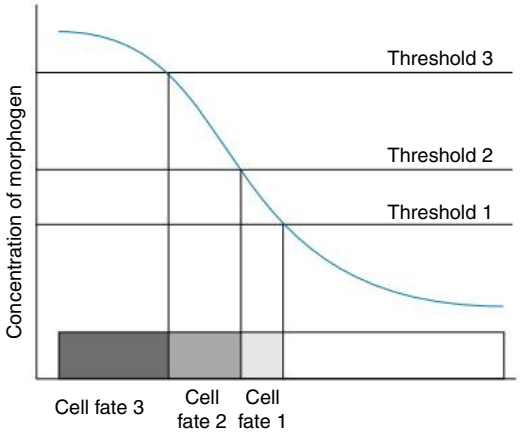
165 The Timing of Development

Key Idea: Embryonic development is regulated by signal molecules that affect gene expression.

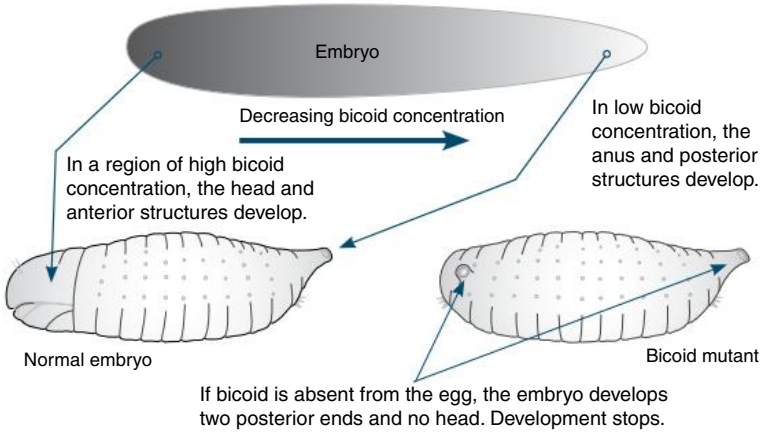
The differentiation of cells is controlled by cell signalling, either by producing a gradient of molecules through the embryo or by signalling a specific cell that then signals other cells in sequence. Cell signals regulate transcription factors that in turn control the differential gene expression that shapes embryos. This process, called **embryonic induction**, takes hours to days, while the time a cell can respond to an inducing signal is strictly limited.

Development in *Drosophila*

Morphogen concentration and cell fate



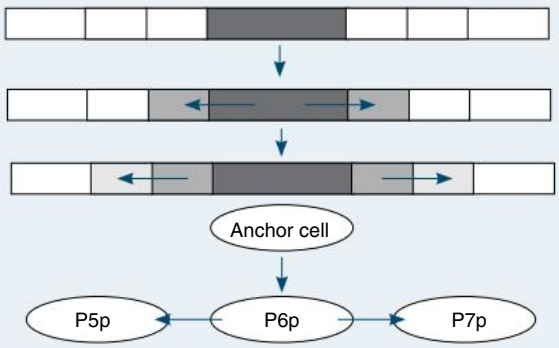
Morphogens are signal molecules that govern patterns of tissue development. Morphogens induce cells in different positions to adopt different fates by diffusing from an area of production through a field of cells.



The **bicoid** gene is important in *Drosophila* development. After fertilisation, bicoid mRNA from the mother is passed to the egg where it is translated into bicoid protein. Bicoid protein forms a concentration gradient in the developing embryo, which determine where the anterior and posterior develop.

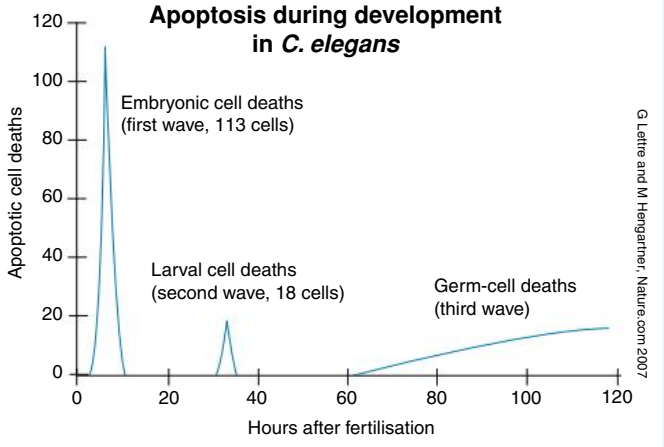
Development in *Caenorhabditis elegans*

A second way of inducing change in embryonic cells is through **sequential induction**. A signal molecule reaches cell A, which responds by developing and producing signal B and so on.



The nematode worm, *Caenorhabditis elegans*, is a **model organism** for developmental studies. The cells that form the vulva (ventral opening for copulation and egg laying) display sequential induction. Signals from the anchor cell induce a change in cell P6p. Cell P6p then signals cells P5p and P7p to develop.

The fate of all of *C. elegans*' 1090 somatic cells are known. 131 somatic cells undergo apoptosis. The death of these cells is controlled by the genes *ced-3* and *ced-4*, which are themselves regulated by gene *ced-9*. There are three waves of apoptosis. The first removes 113 of 628 cells, the second removes 18 more cells, and the third removes half of the developing oocytes.



1. (a) Describe the role of **morphogens**: _____

_____

(b) Explain the purpose of the **bicoid protein** in *Drosophila*: _____

_____

2. Describe **sequential induction** in *C. elegans*: _____

_____

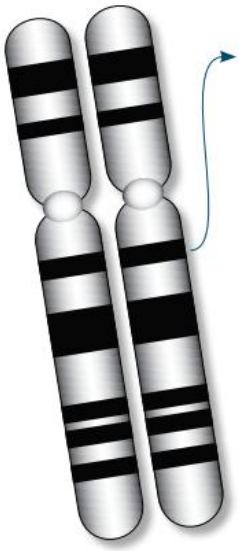
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166 Mutations and Development

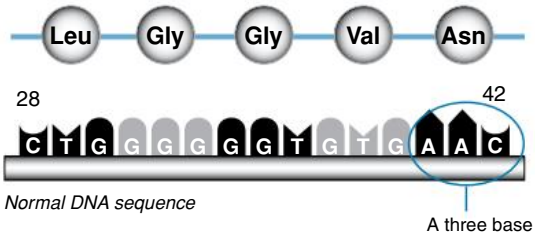
Key Idea: Errors in DNA replication during meiosis can produce new alleles that can be inherited.

Every time DNA is replicated, there is a chance that a copying error will occur. DNA replication has a low **error rate**, with only one mistake for every billion base pairs copied. Changes to the DNA base sequence, called **mutations**, with no effect on the coded product are called silent. However, some mutations can create new alleles. Most new alleles will be detrimental. One example is NSRD, a common form of genetic hearing loss, which accounts for up to 50% of childhood deafness.



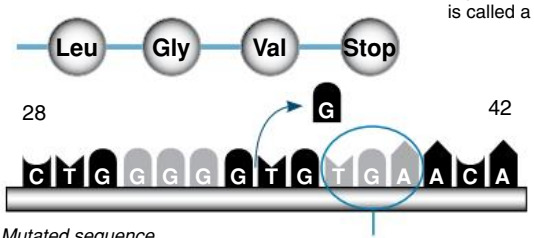
Chromosome 13

The gene that codes for the peptide chain connexin 26.



Normal DNA sequence

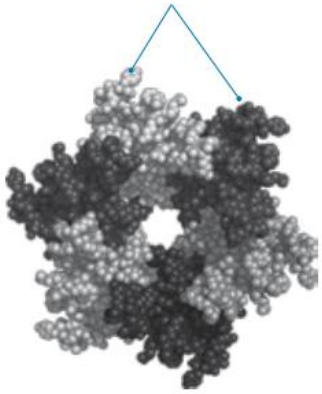
A three base sequence coding for an amino acid is called a **codon**



Mutated sequence

TGA codes for stop

Connexin 26 peptide chains



Six connexin 26 peptide chains join together to form a connection between cells. If these proteins are not made correctly, they are unable to form the final structure.

The most common mutation in this gene is **deletion** of the **35th base** (**G**). DNA is read in groups of three bases, so deleting the guanine alters the reading sequence. The result is a short peptide chain, which cannot function correctly and results in deafness.

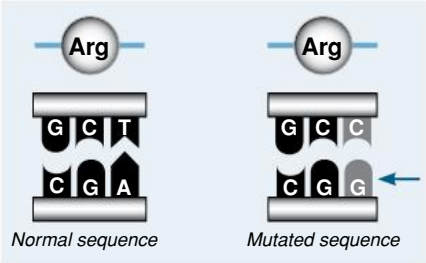
The deletion of this one base causes the formation of a **recessive allele**. Deafness results when there are two recessive alleles.





Harmful Mutations

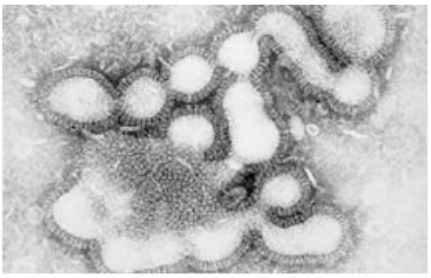
Most mutations cause harmful effects, usually because they stop or alter the production of a protein (often an enzyme). Albinism (above) is one of the more common mutations in nature, and leaves an animal with no pigmentation.



Normal sequence *Mutated sequence*

Silent Mutations

Silent mutations do not change the amino acid sequence nor the final protein. In the genetic code, several codons may code for the same amino acid. Silent mutations are also **neutral** if they do not alter the fitness of the organism.



Beneficial Mutations

Sometimes mutations help the survival of an organism. In viruses (e.g. *Influenzavirus* above) genes coding for the glycoprotein coat are constantly mutating, producing new strains that avoid detection by the host's immune system.

1. How can changes in a DNA sequence occur? _____

2. How can a mutation in a single base be as damaging as a mutation in a sequence of bases? _____

3. Explain how mutation can be harmful or beneficial: _____

KNOW WEB 166 LINK 121 LINK 172 LINK 176

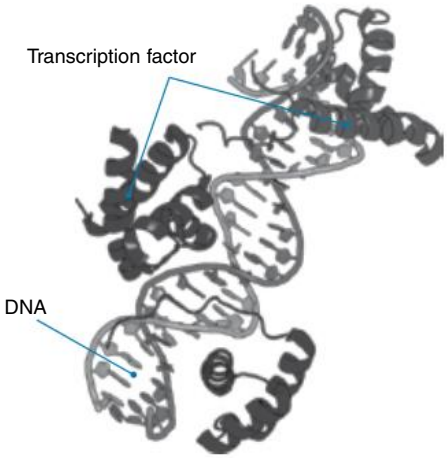
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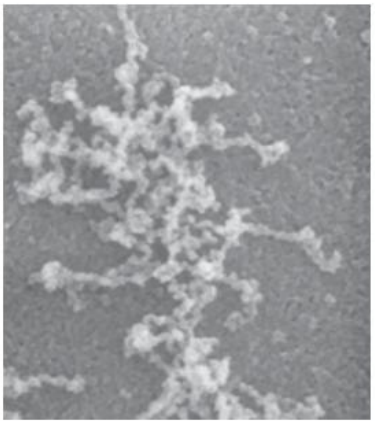
167 Controlling Gene Expression

Key Idea: Gene expression is tightly regulated. It begins when RNA polymerase attaches to the promoter region of a gene.

All the cells in your body contain identical copies of your genetic instructions. Yet these cells appear very different (e.g. muscle, nerve, and epithelial cells have little in common). These morphological differences reflect profound differences in the expression of genes during the cell's development. For example, muscle cells express the genes for the proteins that make up the contractile elements of the muscle fibre. This diversity of cell structure and function reflects the precise control over the time, location, and extent of expression of a huge variety of genes.

The physical state of the DNA in or near a gene is important in helping to control whether the gene is even available for transcription. When the **heterochromatin** is condensed, the transcription proteins cannot reach the DNA and the gene is not expressed. To be transcribed, a gene must first be unpacked from its condensed state. Once unpacked, control of gene expression involves the interaction of **transcription factors** (shown attached to the DNA molecule right) with DNA sequences that control the specific gene. Initiation of transcription is the most important and universally used control point in gene expression.



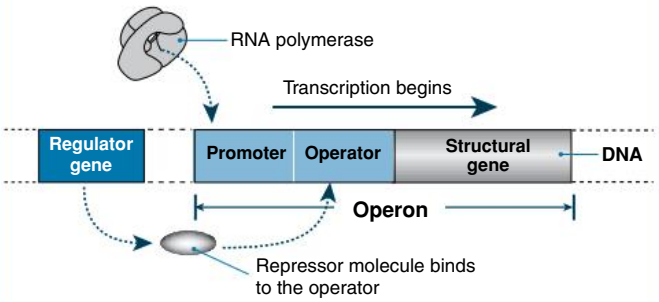


Unravelled mRNA after transcription.

Before transcription can take place, RNA polymerase must attached to the **promoter** region of the DNA. The promoter is upstream of the DNA to be transcribed. The process by which RNA polymerase binds to the promoter is different in prokaryotes and eukaryotes (below).

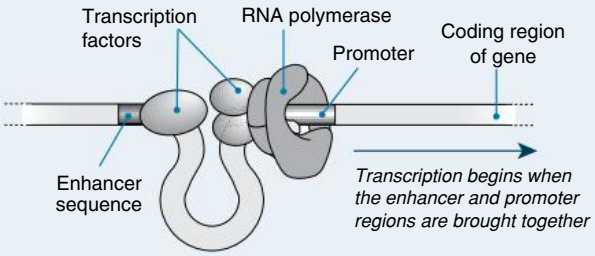
Control of Gene Expression in Prokaryotes

In **prokaryotes**, the RNA polymerase and associated proteins bind directly to the promoter region. The promoter is normally very close to the DNA region to be transcribed. Transcription of the structural gene is controlled by a **regulator** gene, which produces a repressor molecule that may bind to the operator and block transcription. The promoter, operator, and DNA to be transcribed are called an **operon**.



Control of Gene Expression in Eukaryotes

Eukaryote genes do not exist as operons. In **eukaryotes**, several **transcription factors** are required to bind the RNA polymerase to the promoter (the RNA polymerase cannot bind directly). Some of these transcription factors may also bind to the **enhancer** region which may be quite distant from the promoter. The transcription factors cause the promoter and enhancer to come together, which initiates transcription.



1. (a) How is transcription initiated in prokaryotes? _____

_____

_____

_____

(b) How is transcription initiated in eukaryotes? _____

_____

_____

_____

2. What is the role of the promoter? _____

_____

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168 Defective Gene Regulation in Cancer

Key Idea: Cancerous cells have lost their normal cellular control mechanisms. Cancer may be caused by carcinogens. Cells that become damaged beyond repair will normally undergo a programmed cell death (**apoptosis**), which is part of the cell's normal control system. Cancer cells evade this control and become immortal, continuing to divide without any regulation. **Carcinogens** are agents capable of causing cancer. Roughly 90% of carcinogens are also **mutagens**, i.e. they damage DNA. Long-term exposure to carcinogens accelerates the rate at which dividing cells make errors. Any one of a number of cancer-causing factors (including defective genes) may interact to induce cancer.

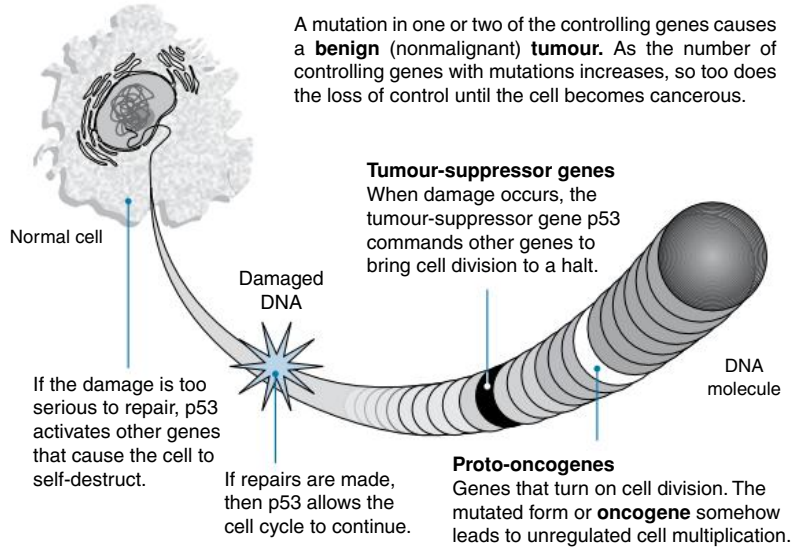
Cancer: Cells out of Control

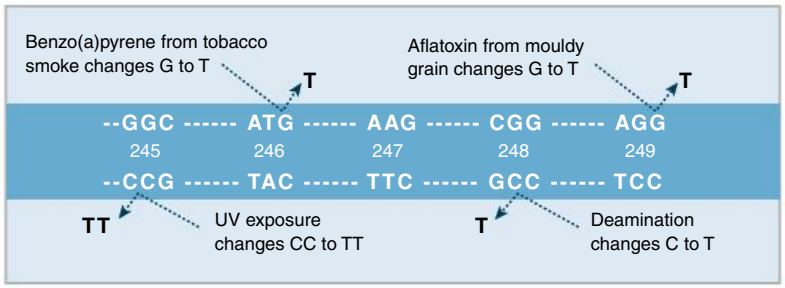
Cancerous transformation results from changes in the genes controlling normal cell growth and division. The resulting cells become immortal and no longer carry out their functional role. Two types of gene are normally involved in controlling the cell cycle: **proto-oncogenes**, which start the cell division process and are essential for normal cell development, and **tumour-suppressor genes**, which switch off cell division. In their normal form, both kinds of genes work as a team, enabling the body to perform vital tasks such as repairing defective cells and replacing dead ones. But mutations in these genes can disrupt these finely tuned checks and balances. Proto-oncogenes, through mutation, can give rise to **oncogenes**; genes that lead to uncontrollable cell division. Mutations to tumour-suppressor genes initiate most human cancers. The best studied tumour-suppressor gene is **p53**, which encodes a protein that halts the cell cycle so that DNA can be repaired before division.

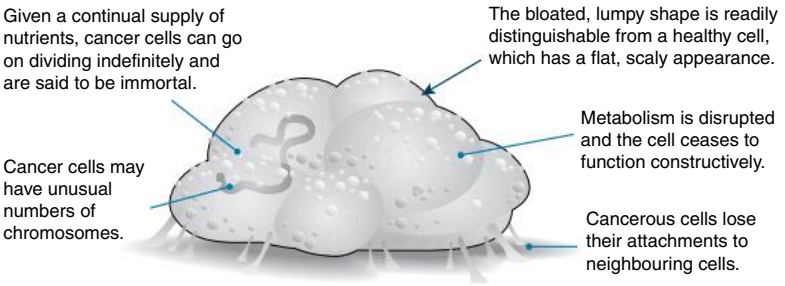
The panel, right, shows the mutagenic action of some selected carcinogens on four of five codons of the **p53 gene**.

Features of Cancer Cells

The diagram right shows a single **lung cell** that has become cancerous. It no longer carries out the role of a lung cell, and instead takes on a parasitic lifestyle, taking nutrients from the body and contributing nothing in return. The rate of cell division is greater than in normal cells in the same tissue because there is no *resting phase* between divisions.







1. How do cancerous cells differ from normal cells? _____

_____

2. Explain how the cell cycle is normally controlled, including reference to the role of **tumour-suppressor genes**:

_____

_____

_____

_____

3. With reference to the role of **oncogenes**, explain how the normal controls over the cell cycle can be lost:

_____

_____

_____

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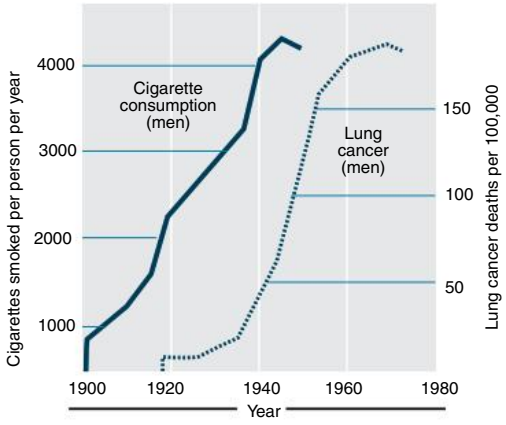
169 KEY TERMS: Did You Get It?

1. Test your vocabulary by matching each term to its definition, as identified by its preceding letter code.

adult stem cells	**A** The entire process of transferring of information encoded in a gene into a gene product.
cancer	**B** Unspecialised cells that are able to differentiate into any one of many different kinds of cell.
cellular differentiation	**C** Developmental control genes that are involved in establishing the anterior-posterior axis of an organism. They are shared by almost all organisms.
embryonic stem cells	**D** Pluripotent stem cells derived from the inner cell mass of the blastocyst (early-stage embryo).
gene expression	**E** Undifferentiated, multipotent cells found in tissues or organs that may give rise to several cell types, but are primarily involved in the maintenance and repair of the tissue in which they are found.
Hox genes	**F** Process by which a less specialised cell becomes more specialised in order to perform specific function.
morphogens	**G** A change to the DNA sequence of an organism. This may be a deletion, insertion, duplication, inversion or translocation of DNA in a gene or chromosome.
mutation	**H** Signal molecules that control and direct the development of cells and tissue by producing a concentration gradient.
stem cells	**I** Disease condition in which mutations in the genes regulating cell growth and division allow cells to grow and divide in an uncontrolled fashion, resulting in a tumour.

2. In relation to developing and maintaining shape and form, explain why organisms need to tightly control gene expression.

3. A study aimed to determine if there is a correlation between smoking and lung cancer. Analyse the graph (right) and state whether or not you think there is a correlation. Explain your answer:



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QLD As required
VCE Unit 4 (1)
WA ATAR Unit 3
SA Theme O

Inheritance

Key terms

allele
aneuploidy
carrier
codominance (of alleles)
continuous variation
diploid
dihybrid cross
discontinuous variation
dominant (of alleles)
F_1 / F_2 generation
genomic imprinting
genotype
haploid
heterozygous
homologous chromosomes
homozygous
independent assortment
linkage
locus
meiosis
monohybrid cross
multiple alleles
mutation
pedigree analysis
phenotype
polygenes
Punnett square
pure (true)-breeding
recessive (of alleles)
recombination
sex chromosome
sex linked gene
test cross
trait
trisomy

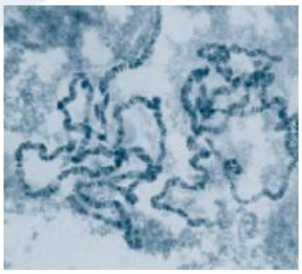
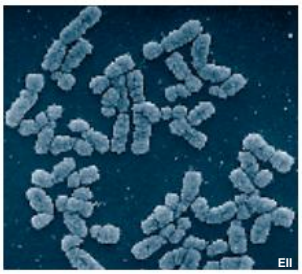
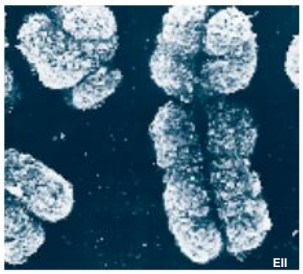


Sources of variation

Learning aims and skills

Activity number

- ☐ 1 Identify and describe sources of variation in sexually reproducing organisms. Recall how linkage and recombination affect the inheritance of alleles. — 169
- ☐ 2 Explain how genotype and environment contribute to phenotypic variation. — 170
- ☐ 3 Explain how gene mutations, e.g. sickle cell mutation and cystic fibrosis, contribute to phenotypic variation. — 171-173
- ☐ 4 Describe chromosome mutations and explain their evolutionary significance. — 174 175
- ☐ 5 Describe and explain aneuploidy arising as a result of non-disjunction during meiosis. Recognise Down syndrome (trisomy 21) as a type of aneuploidy. — 176 177
- ☐ 6 Explain the basis of sex determination in humans (XX-female, XY-male). Explain how the environment may determine sex in some animals. — 196

Studying inheritance

Learning aims and skills

Activity number

- ☐ 7 Summarise Mendel's principles of inheritance and their importance to our understanding of heredity and evolution. — 178 179
- ☐ 8 Demonstrate appropriate use of the terms used in studying inheritance: allele, locus, trait, heterozygous, homozygous, genotype, phenotype, cross, test cross, back cross, carrier, F_1/F_2 generation. — 180 181
- ☐ 9 Use a Punnett square to solve problems involving monohybrid inheritance with a simple dominant-recessive pattern. — 181-184
- ☐ 10 Describe and explain inheritance involving codominance in multiple alleles, sex linked genes, and (if required) lethal alleles. — 185-187 197-198
- ☐ 11 Use a Punnett square to solve problems involving dihybrid inheritance of unlinked, autosomal genes for two independent characteristics. Use the test cross to determine an unknown dominant genotype. — 188 193
- ☐ 12 Use the chi-squared test to test the significance of differences between the observed and expected results of genetic crosses. — Weblink
- ☐ 13 EXTENSION: Explain how some traits are the result of non-nuclear inheritance and describe an example. Describe and explain genomic imprinting. — 195 196
- ☐ 14 Explain the use of pedigree analysis to illustrate the inheritance of traits — 200

Gene interactions

Learning aims and skills

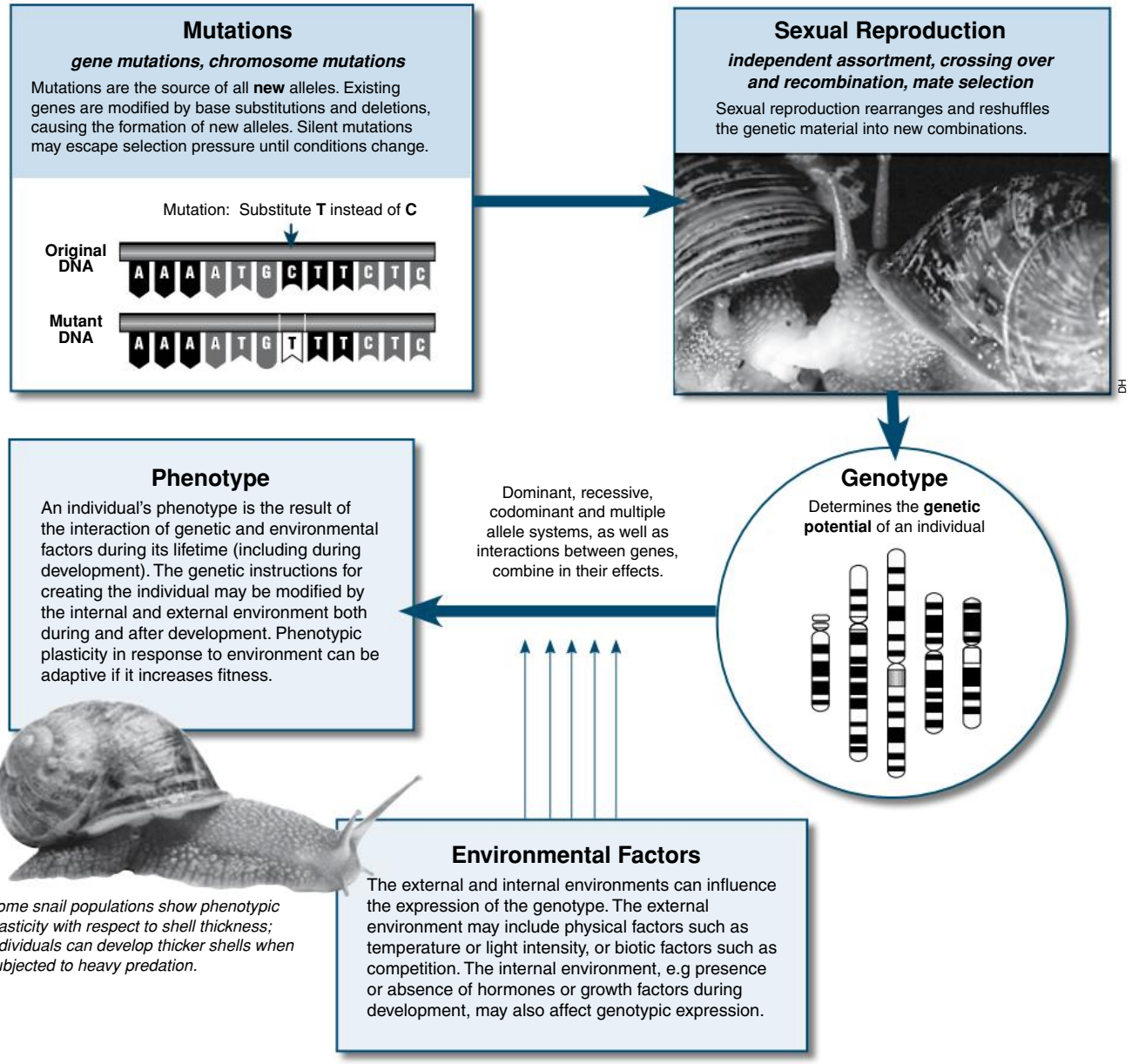
Activity number

- ☐ 15 Describe and explain polygenic inheritance (polygenes are also called multiple genes). Distinguish between continuous and discontinuous variation in phenotypes. Explain the genetic basis for each pattern. — 199

170 Sources of Variation

Key Idea: Variation within populations allows for adaptation by natural selection to changes in the environment.

Variation refers to the diversity of genetic and phenotypic traits within and between populations. Variation is a feature of sexually reproducing populations and it is the essential raw material for selection. Variation gives species greater opportunity to adapt to and survive in a dynamic environment. Populations of highly variable species will include individuals with different **fitness** in the prevailing environment. It is this variability that offers the chance of reproductive advantage if environments change. Species with low variability (such as populations of clones) can be highly successful in stable conditions, but will reproduce sexually when environments become unfavourable for just this reason. Phenotype is influenced by both genotype and the environment. Even organisms of fixed genotype may differ in how much their phenotype can change in response to environment (a phenomenon known as phenotypic plasticity).



Some snail populations show phenotypic plasticity with respect to shell thickness; individuals can develop thicker shells when subjected to heavy predation.

1. Using examples, explain how the environment of a particular genotype can affect the phenotype: _____

2. Discuss the significance of variation in selection: _____

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LINK 215 LINK 171 LINK 136 KNOW



Albinism (above) is the result of the inheritance of recessive alleles for melanin production. Those with the albino phenotype lack melanin pigment in the eyes, skin, and hair.

Comb shape in poultry is a **qualitative trait** and birds have one of four phenotypes depending on which combination of four alleles they inherit. The dash (missing allele) indicates that the allele may be recessive or dominant.







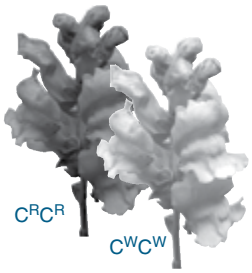


Quantitative traits are characterised by **continuous variation**, with individuals falling somewhere on a normal distribution curve of the phenotypic range. Typical examples include skin colour and height in humans (left), grain yield in corn (above), growth in pigs (above, left), and milk production in cattle (far left). Quantitative traits are determined by genes at many loci (polygenic) but most are also influenced by environmental factors.



Single comb **rrpp** | Walnut comb **R_P_** | Pea comb **rrP_** | Rose comb **R_pp**

Flower colour in snapdragons (right) is also a **qualitative trait** determined by two alleles. (red and white) The alleles show incomplete dominance and the heterozygote (C^RC^W) exhibits an intermediate phenotype between the two homozygotes.



3. Describe three ways in which sexual reproduction can provide genetic variation in individuals:

   (a) _____

   _____

   (b) _____

   _____

   (c) _____

   _____

4. (a) What is a neutral mutation? _____

   (b) What is the significance of neutral mutations? _____

   _____

5. Describe the differences between **continuous** and **discontinuous** variation, giving examples to illustrate your answer:

   _____

   _____

   _____

6. Identify each of the following phenotypic traits as continuous (quantitative) or discontinuous (qualitative):

   (a) Wool production in sheep: _____ (d) Albinism in mammals: _____

   (b) Hand span in humans: _____ (e) Body weight in mice: _____

   (c) Blood groups in humans: _____ (f) Flower colour in snapdragons: _____

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171 Gene-Environment Interactions

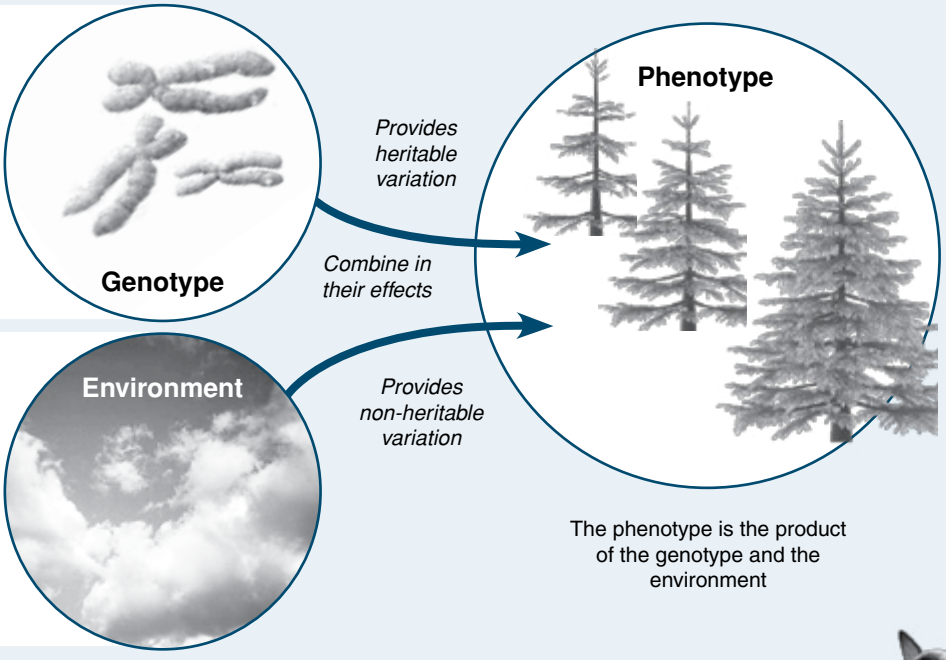
Key Idea: An organism's phenotype is influenced by the environment in which it develops, even though the genotype remains unaffected.

External environmental factors can modify the phenotype encoded by genes. This can occur both during development and later in life. Even identical twins have minor differences in their appearance due to environmental factors such as diet and intrauterine environment before birth. Environmental factors that affect the phenotype include nutrients or diet, temperature, and the presence of other organisms.

Sources of Variation in Organisms

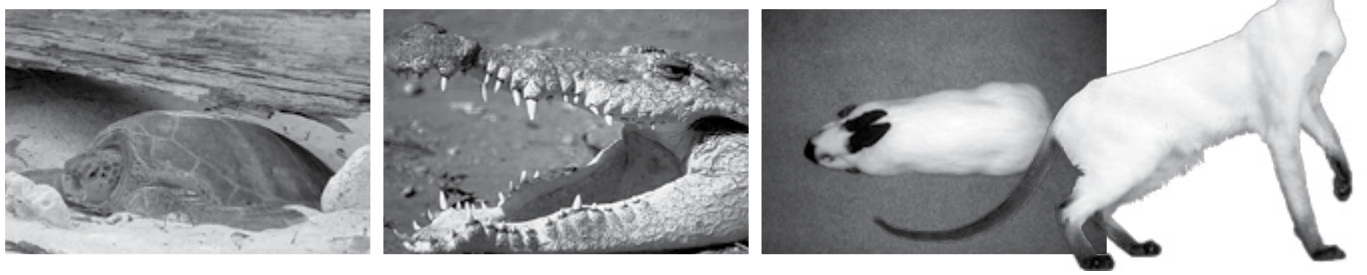
Sources of genetic variation
- Dominant alleles
- Recessive alleles
- Mutations
- Crossing over
- Independent assortment
- Gene interactions

Sources of environmentally induced variation
- Climate and altitude
- Light or water availability
- Acidity (pH)
- Soil type or nutrition
- Presence of toxins
- Predation or disease
- Competition



The phenotype is the product of the genotype and the environment

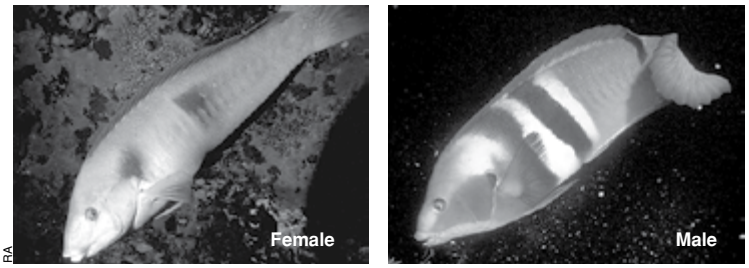
The Effect of Temperature



The sex of some animals is determined by the incubation temperature during their embryonic development. Examples include turtles, crocodiles, and the American alligator. In some species, high incubation temperatures produce males and low temperatures produce females. In other species, the opposite is true. Temperature regulated sex determination may be advantageous by preventing inbreeding (since all siblings will tend to be of the same sex).

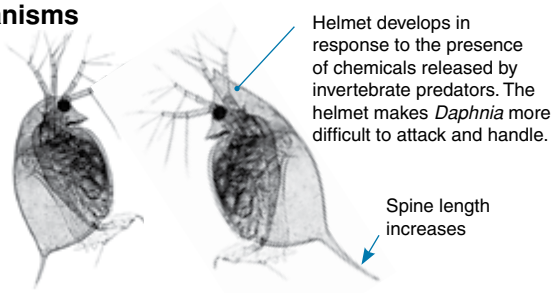
Colour-pointing in breeds of cats and rabbits (e.g. Siamese, Himalayan) is a result of a temperature sensitive mutation in one of the enzymes in the metabolic pathway from tyrosine to melanin. The dark pigment is only produced in the cooler areas of the body (face, ears, feet, and tail), while the rest of the body is a paler version of the same colour, or white.

The Effect of Other Organisms



RA

The presence of other individuals of the same species may control sex determination for some animals. Some fish species, including some in the wrasse family (e.g. *Coris sandageri*, above), show this phenomenon. The fish live in groups consisting of a single male with attendant females and juveniles. In the presence of a male, all juvenile fish of this species grow into females. When the male dies, the dominant female will undergo physiological changes to become a male. The male has distinctive bands, whereas the female is pale in colour and has very faint markings.

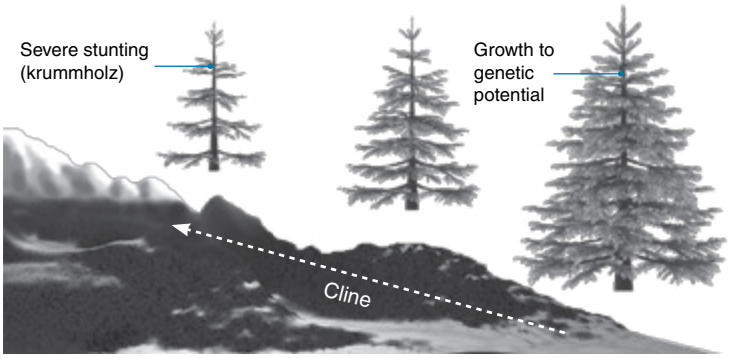


Some organisms respond to the presence of other, potentially harmful, organisms by changing their morphology or body shape. Invertebrates such as *Daphnia* will grow a large helmet when a predatory midge larva is present. Such responses are usually mediated through chemicals produced by the predator (or competitor), and are common in plants as well as animals.

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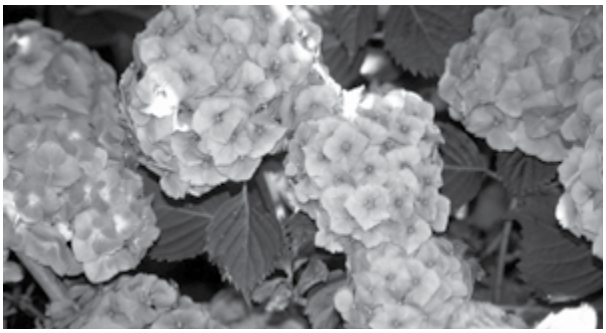
LINK 218 LINK 217 LINK 195 KNOW

The Effect of Altitude



Increasing altitude can stunt the phenotype of plants with the same genotype. In some conifers, e.g. Engelmann spruce, plants at low altitude grow to their full genetic potential, but become progressively more stunted as elevation increases, forming krummholz (gnarled bushy growth forms) at the highest, most severe sites. A continuous gradation in a phenotypic character within a species, associated with a change in an environmental variable, is called a **cline**.

The Effect of Chemical Environment



The chemical environment can influence the expressed phenotype in both plants and animals. In hydrangeas, flower colour varies according to soil pH. Flowers are blue in more acid soils (pH 5.0-5.5), but pink in more alkaline soils (pH 6.0-6.5). The blue colour is due to the presence of aluminium compounds in the flowers and aluminium is more readily available when the soil pH is low.

1. Describe an example to illustrate how genotype and environment contribute to phenotype: _____

2. What are the physical factors associated with altitude that could affect plant phenotype? _____

3. Describe an example of how the chemical environment of a plant can influence phenotype: _____

4. Why are the darker patches of fur in colour-pointed cats and rabbits found only on the face, paws and tail:

5. There has been much amusement over the size of record-breaking vegetables, such as enormous pumpkins, produced for competitions. How could you improve the chance that a vegetable would reach its maximum genetic potential?

6. (a) What is a **cline**? _____

   (b) On a windswept portion of a coast, two different species of plant (species A and species B) were found growing together. Both had a low growing (prostrate) phenotype. One of each plant type was transferred to a greenhouse where "ideal" conditions were provided to allow maximum growth. In this controlled environment, species B continued to grow in its original prostrate form, but species A changed its growing pattern and became erect in form. Identify the **cause** of the prostrate phenotype in each of the coastal grown plant species and explain your answer:

   Plant species A: _____

   Plant species B: _____

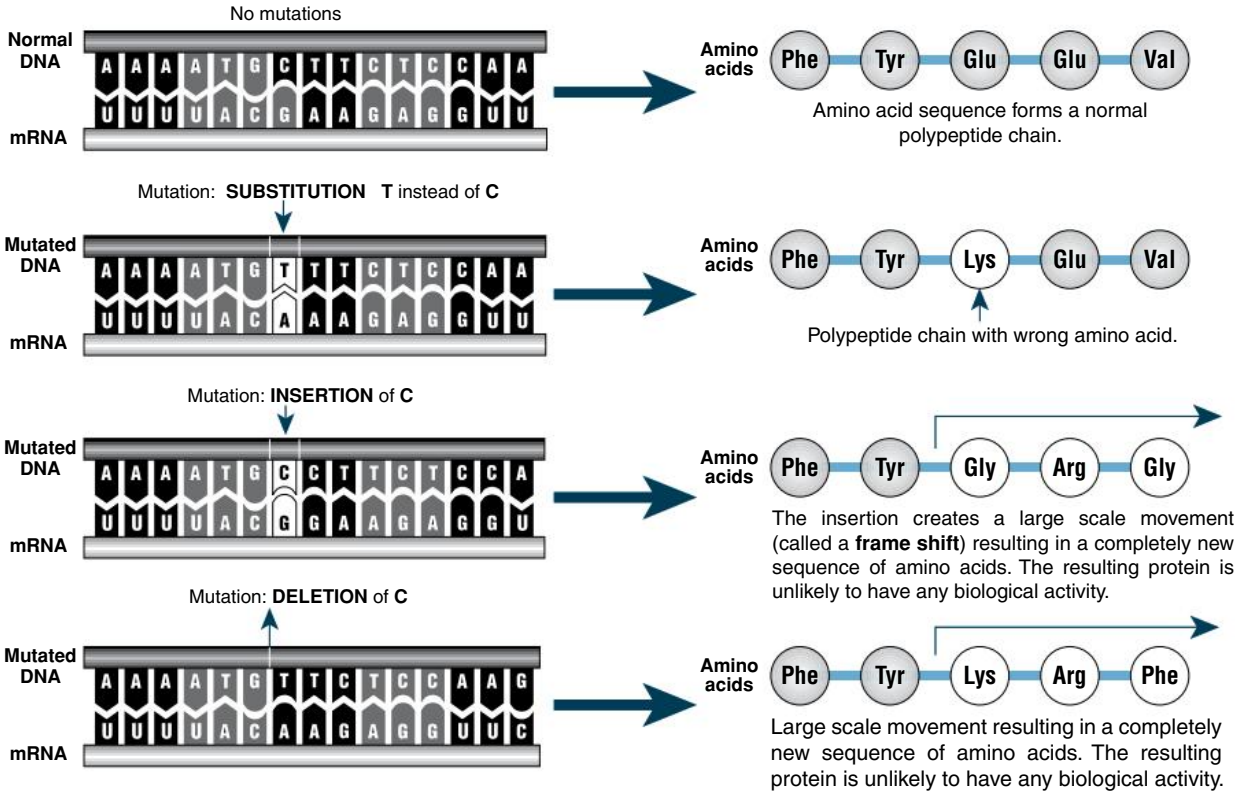
   (c) Which of these species (A or B) would be most likely to exhibit clinal variation? _____

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172 Gene Mutations

Key Idea: Gene mutations are localised changes to the DNA base sequence.

Gene mutations are small, localised changes in the base sequence of a DNA strand caused by a mutagen or an error during DNA replication. The changes may involve a single nucleotide (a **point mutation**) or a change to a triplet. Point mutations can occur by substitution, insertion, or deletion of bases. These changes alter the mRNA transcribed from the mutated DNA. A point mutation may not alter the amino acid sequence because more than one codon can code for the same amino acid. Mutations that result in a change in the amino acid sequence will most often be harmful.



1. Explain what is meant by a **frame shift mutation**: _____

_____

2. Some gene mutations are more disruptive to an organism than others.

   (a) Identify which type of gene mutations are the most damaging to an organism: _____

   (b) Explain why they are the most disruptive: _____

   _____

   (c) Describe what type of gene mutation is least likely to cause a change in protein structure and explain your answer:

   _____

   _____

3. In the following DNA sequence, replace the **G** of the **second codon** with an A to create a new mutant DNA, then determine the new mRNA sequence, and the amino acid sequence. Refer to the mRNA-amino acid table to identify the amino acids coded in each case.

   (a) Original DNA: AAA ATG TTT CTC CAA GAT

   Mutated DNA: _____

   mRNA: _____

   Amino acids: _____

   (b) Identify the amino acid coded by codon 2 (ATG) in the original DNA: _____

   (c) Explain the effect of the mutation: _____

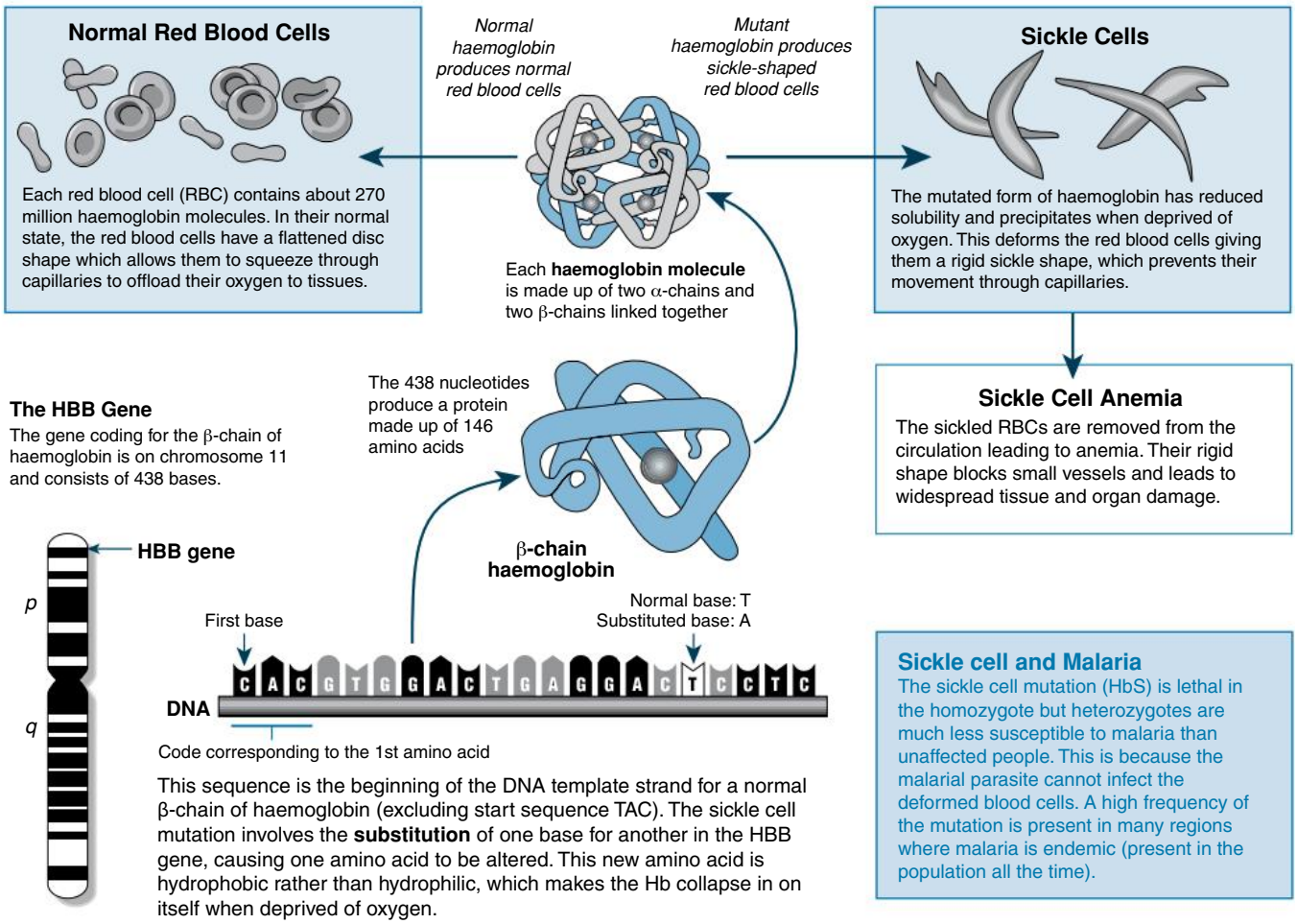
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LINK 174 LINK 173 LINK 166 LINK 121 WEB 172 KNOW

173 Sickle Cell Mutation

Key Idea: The substitution of one nucleotide from T to A results in sickle cell disease. The mutation is codominant.

Sickle cell disease is an inherited blood disorder caused by a gene mutation (Hb^S), which produces a faulty beta (β) chain haemoglobin (Hb) protein. This in turn produces red blood cells with a deformed sickle appearance and a reduced capacity to carry oxygen. Many aspects of metabolism are also affected. The mutation is codominant (both alleles equally expressed), and people heterozygous for the mutation (carriers) have enough functional haemoglobin and suffer only minor effects.



1. For the DNA sequence in the diagram above, state the number of:

   (a) Bases: _____ (b) Triplets: _____ (c) Amino acids coded for: _____

2. (a) Explain the genetic cause of sickle cell disease: _____

   _____

   (b) How does the sickle cell mutation result in the symptoms of the disease? _____

   _____

   _____

   (c) Explain why heterozygotes (carriers) suffer only minor effects: _____

   _____

   _____

   (d) Carriers show symptoms of the disease if they are deprived of oxygen (for example, at altitude). Suggest why:

   _____

   _____

3. Briefly explain why there is a high frequency of the sickle cell mutation in populations where malaria is endemic:

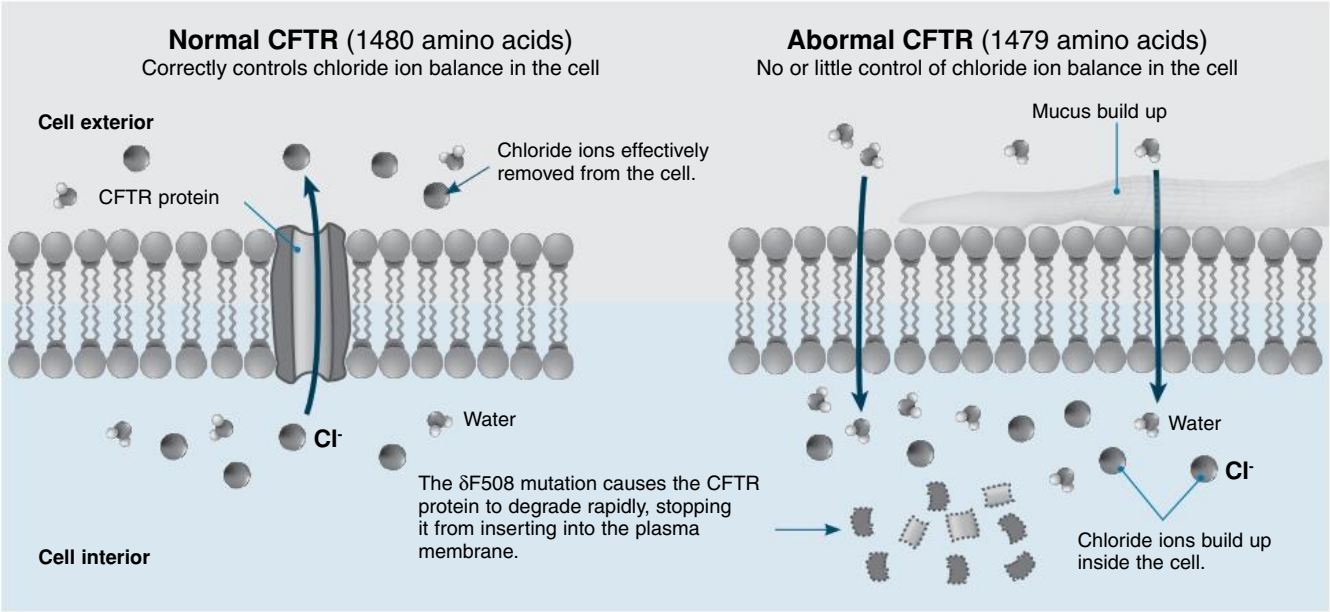
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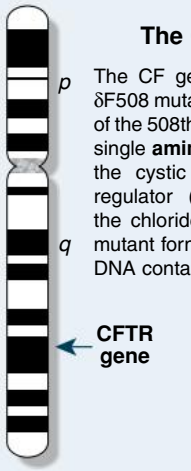
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174 Cystic Fibrosis Mutation

Key Idea: Cystic fibrosis most often results from a triplet deletion in the CFTR gene, producing a protein that is unable to regulate chloride transport.

Cystic fibrosis (CF) is an inherited disorder caused by a mutation of the CFTR gene. It is one of the most common lethal autosomal recessive conditions affecting people of European descent (4% are carriers). The CFTR gene's protein product is a membrane-based protein that regulates chloride transport in cells. Over 500 mutations of the CFTR gene are known, causing disease symptoms of varying severity. The δ(delta)F508 mutation accounts for more than 70% of all defective CFTR genes. This mutation leads to an abnormal CFTR, which cannot take its proper position in the membrane (below) nor perform its transport function.





The CF gene on chromosome 7

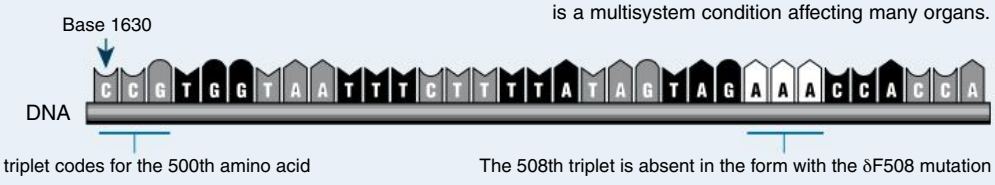
The CF gene is located on chromosome 7. The δF508 mutation of the CF gene describes a deletion of the 508th triplet, which in turn causes the loss of a single **amino acid** from the gene's protein product, the cystic fibrosis transmembrane conductance regulator (CFTR). This protein normally regulates the chloride channels in cell membranes, but the mutant form fails to achieve this. The portion of the DNA containing the mutation site is shown below:



The CFTR protein consists of 1480 amino acids

CFTR protein

The δF508 mutant form of CFTR fails to take up its position in the membrane. Its absence results in defective chloride transport and leads to a net increase in water absorption by the cell. This accounts for the symptoms of cystic fibrosis, where mucus-secreting glands, particularly in the lungs and pancreas, become fibrous and produce abnormally thick mucus. The widespread presence of CFTR throughout the body also explains why CF is a multisystem condition affecting many organs.



1. (a) Write the mRNA sequence for the transcribing DNA strand above: _____

_____

(b) Use the mRNA-amino acid table earlier in this workbook to determine the amino acid sequence coded by the mRNA for the fragment of the normal protein we are studying here:

_____

2. (a) Rewrite the mRNA sequence for the mutant DNA strand: _____

_____

(b) Determine the amino acid sequence coded by the mRNA for the fragment of the δF508 mutant protein:

_____

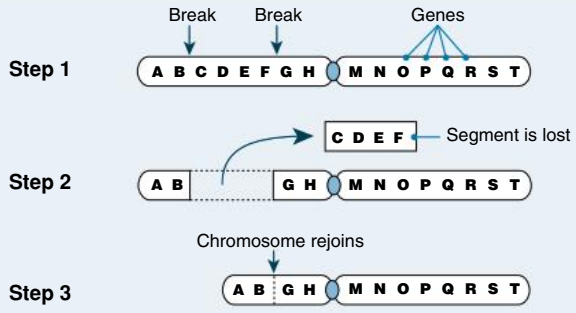
(c) Which amino acid has been removed from the protein by this mutation? _____

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175 Chromosome Mutations

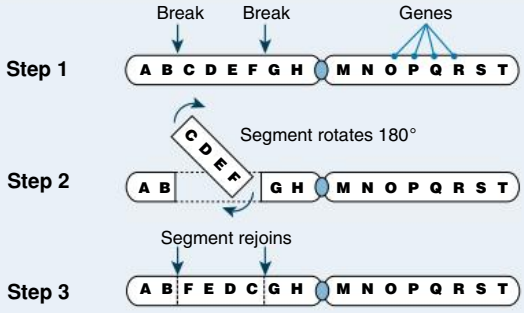
Key Idea: Large scale mutations occurring during meiosis can fundamentally change chromosome structure.

Chromosome mutations (also called block mutations) involve the rearrangement of whole blocks of genes, rather than individual bases within a gene. They commonly occur during meiosis and they alter the number or sequence of whole sets of genes on the chromosome (represented by letters below). Translocations may sometimes involve the fusion of whole chromosomes, thereby reducing the chromosome number of an organism. This is thought to be an important evolutionary mechanism by which instant speciation can occur.



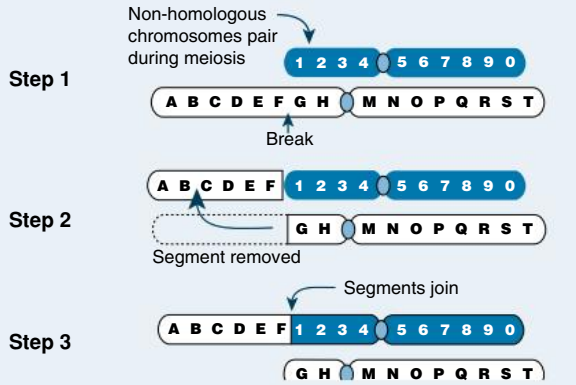
Deletion

A break may occur at two points on the chromosome and the middle piece of the chromosome falls out. The two ends then rejoin to form a chromosome deficient in some genes. Alternatively, the end of a chromosome may break off and is lost.



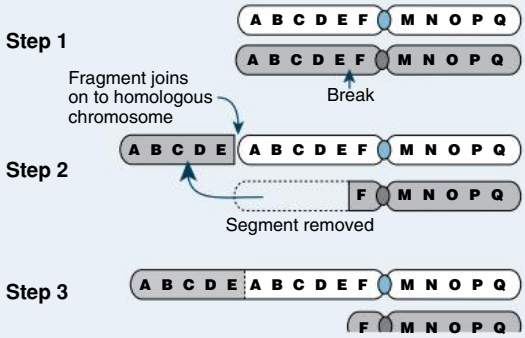
Inversion

The middle piece of the chromosome falls out and rotates through 180° and then rejoins. There is no loss of genetic material. The genes will be in a reverse order for this segment of the chromosome.



Translocation

Translocation involves the movement of a group of genes between different chromosomes. The large chromosome (white) and the small chromosome (blue) are not homologous. A piece of one chromosome breaks off and joins onto another chromosome. When the chromosomes are passed to gametes some will receive extra genes, while some will be deficient.



Duplication

A segment is lost from one chromosome and is added to its homologue. In this diagram, the darker chromosome on the bottom is the 'donor' of the duplicated piece of chromosome. The chromosome with the segment removed is deficient in genes. Some gametes will receive double the genes while others will have no genes for the affected segment.

1. For each of the chromosome (block) mutations illustrated above, write the original gene sequence and the new gene sequence after the mutation has occurred (the first one has been done for you):

	Original sequence(s)	Mutated sequence(s)
(a) Deletion:	A B C D E F G H M N O P Q R S T	A B G H M N O P Q R S T
(b) Inversion:		
(c) Translocation:		
(d) Duplication:		

2. Which type of block mutation is likely to be the least damaging to the organism? Explain your answer:

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176 Gene Duplication and Evolution

Key Idea: Gene duplication can provide new genes for natural selection to act on.

Over the last 40 or so years, the role of gene duplication in evolution has been hypothesised. Duplication creates more genetic information, which can then be influenced by selection pressures. Studies have shown that gene duplication has occurred in virtually every species and often more than once. It is well known that many important crop plants (e.g. wheat) have duplicated not just one gene, but their entire genome. The duplication of a gene frees one of those genes to develop a new function, while the original continues as it was. In the case of genes that produce proteins with a tendency or ability to perform two functions, there may be **adaptive conflict**, in which its ability to perform one function compromises its ability to perform another. Gene duplication solves this problem by allowing natural selection to act on the genes so that they follow different evolutionary paths.



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Gene duplication in colobine monkeys has enabled the production of enzymes that optimally perform similar functions in different body environments. The primary food source of colobines, unlike most other primates, is leaves. The leaves are fermented in the gut by bacteria, which are then digested with the assistance of an enzyme produced by RNase genes.

In colobines there are two forms of the RNase genes, **RNase1** and **RNase1B**, while in other primates there is only RNase1.

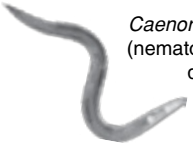
The optimal pH for the enzyme RNase1 is 7.4. For RNase1B, the optimal pH is 6.3. In colobine monkeys, the pH of the digestive system is 6-7, but in other primates it is 7.4-8. RNase1B is six times more efficient at degrading RNA in the gut of colobines than RNase1. RNase1 is also expressed in cells outside the digestive system where it degrades double stranded RNA and may assist in defense against viral infection. RNase1B is 300 times less efficient at this function.



AFP III

Fish living in the near freezing waters of the Antarctic must have a way of ensuring their blood remains ice free. In many species, this is done by employing proteins with antifreeze properties. There are four major antifreeze proteins found in fish (labelled AFP types I - IV). The gene for the protein AFP III, found in Antarctic eelpout, is very similar to the gene that produces sialic acid synthase (SAS) (also found in humans). Molecular studies have found that a slight modification in the SAS gene causes the production and secretion of AFP III. More importantly, the SAS gene also shows ice binding capabilities. It appears that a duplication of the SAS gene produced a new gene that was selected for its ice binding capabilities and thus diverged to become the AFP III gene in Antarctic eelpout.

Gene Duplication



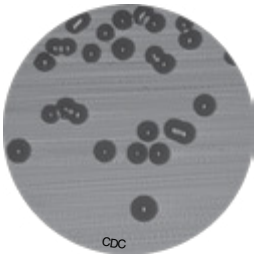
Caenorhabditis elegans (nematode): 49% genes duplicated



Drosophila melanogaster (fruit fly): 41% genes duplicated



Saccharomyces cerevisiae (yeast): 30% genes duplicated



CDC

Haemophilus influenza (bacterium): 17% genes duplicated



Homo sapiens: 38% genes duplicated

	Total gene number	Number of duplicated genes (%)
Bacteria		
Mycoplasma pneumoniae	677	298 (44)
Helicobacter pylori	1590	266 (17)
Archaea		
Archaeoglobus fulgidus	2436	719 (30)
Eukarya		
Arabidopsis thaliana (thale cress)	25,498	16,574 (65)

Adapted from J. Zhang, University Michigan 2003

1. (a) Use an example to explain how gene duplication allows for the evolution of new gene function:

(b) Explain how duplication can resolve adaptive conflict:

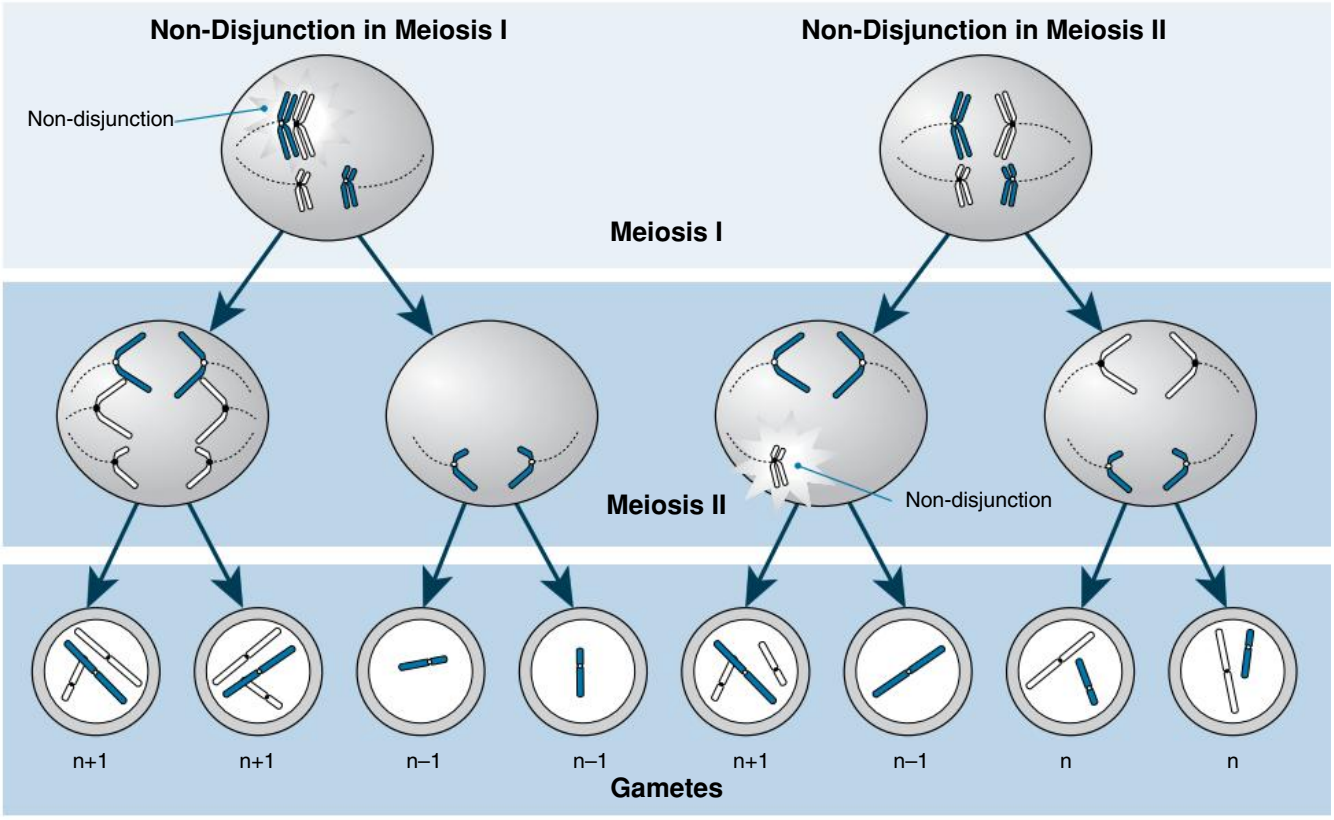
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LINK 175 KNOW

177 Non-Disjunction in Meiosis

Key Idea: Non-disjunction during meiosis results in incorrect apportioning of chromosomes to the gametes.

In meiosis, chromosomes are usually distributed the daughter cells without error. Occasionally, homologous chromosomes fail to separate properly in meiosis I, or sister chromatids fail to separate in meiosis II. In these cases, one gamete receives two of the same type of chromosome and the other gamete receives no copy. This error is known as **non-disjunction** and it results in abnormal numbers of chromosomes in the gametes. The union of an aberrant and a normal gamete at fertilisation produces offspring with an abnormal chromosome number. This condition is known as aneuploidy.





Down Syndrome (Trisomy 21)

Down syndrome is the most common of the human aneuploidies. The incidence rate in humans is about 1 in 800 births for women aged 30 to 31 years, with a **maternal age effect** (the rate increases rapidly with maternal age).

Nearly all cases (approximately 95%) result from **non-disjunction** of chromosome 21 during **meiosis**. When this happens, a gamete (most commonly the oocyte) ends up with 24 rather than 23 chromosomes, and fertilisation produces a trisomic offspring.

Left: Down syndrome phenotype.

Right: A karyogram for an individual with trisomy 21. The chromosomes are circled.

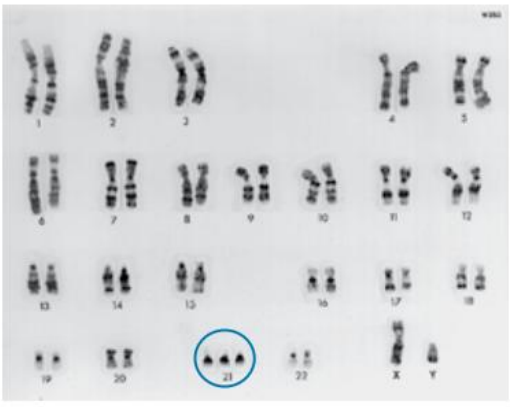


Photo: Waikato Hospital

1. Describe the consequences of non-disjunction during meiosis: _____

_____

_____

2. Explain why non-disjunction in meiosis I results in a higher proportion of faulty gametes than non-disjunction in meiosis II:

_____

_____

3. What is the maternal age effect and what are its consequences? _____

_____

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178 Aneuploidy in Humans

Key Idea: Nondisjunction during meiosis can result in more or less than the normal number of chromosomes.

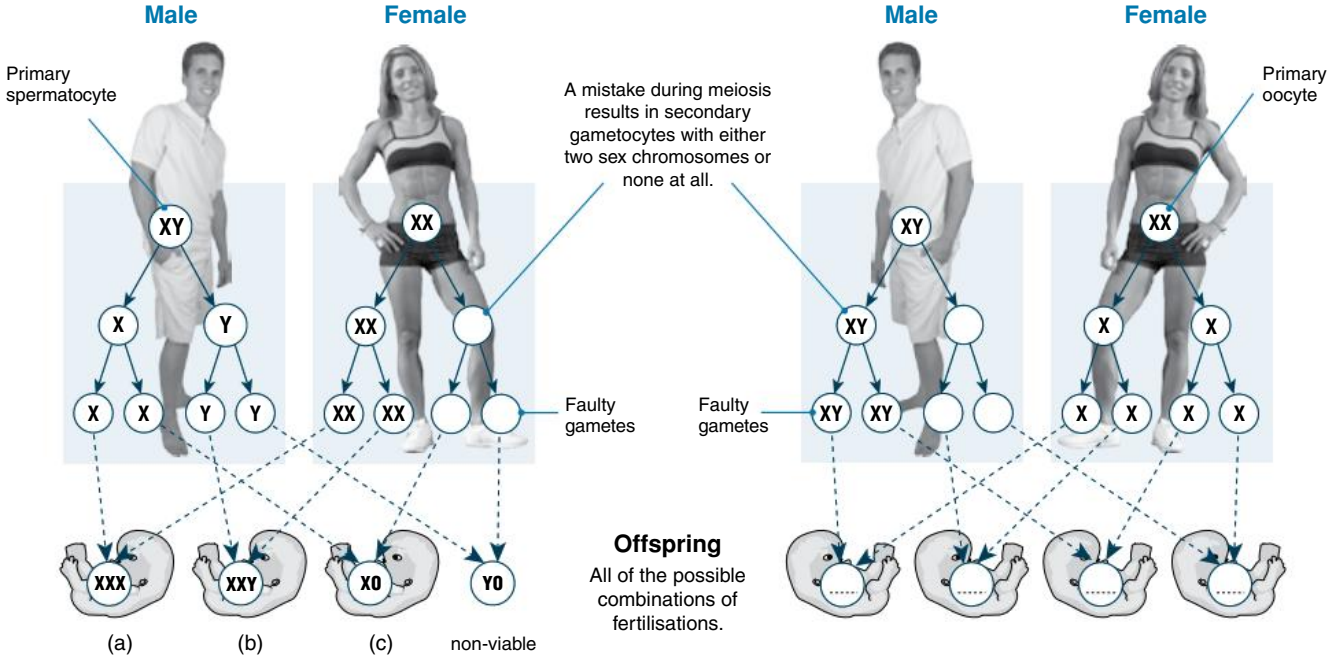
Euploidy is the condition of having an exact multiple of the haploid number of chromosomes. Normal euploid humans have 46 chromosomes (2N). **Aneuploidy** is the condition where the chromosome number is not an exact multiple of the normal haploid set for the species (e.g. 2N+2, 2N–1). Polysomy is aneuploidy involving reduplication of some of the chromosomes beyond the normal diploid number (e.g. 2N+1). Aneuploidy usually results from non-disjunction. The two most common forms are monosomy (e.g. Turner syndrome) and **trisomy** (e.g. Down and Klinefelter syndrome).

Faulty Egg Production

The male has produced normal gametes, but the female has not. The two X-sex chromosomes failed to separate during the first division of meiosis.

Faulty Sperm Production

The female has produced normal gametes, while the male has had an error during the first division of meiosis. The two sex chromosomes (**X** and **Y**) failed to separate during the first division.



1. Identify the sex chromosomes in each of the unlabelled embryos (above, right):

2. Using the table on the next page, identify the syndrome for each of the offspring labelled (a) to (c) above:

   (a) _____ (b) _____ (c) _____

3. Why is the YO configuration (above) non-viable (i.e. there is no embryonic development)? _____

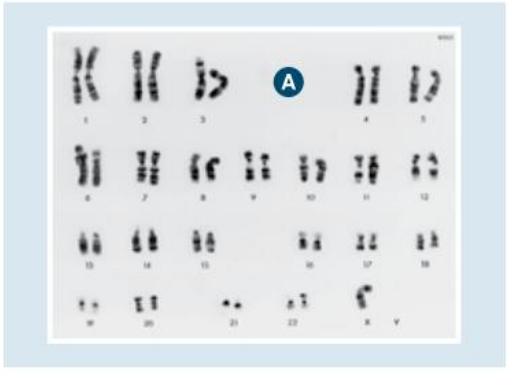
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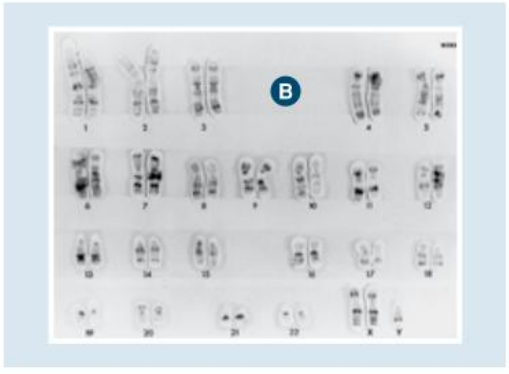
4. (a) For karyotype **A**, below, circle the sex chromosomes and state:

   Chromosome configuration: _____ Sex of individual (M/F): ___ Syndrome: _____

   (b) For karyotype **B**, below, circle the sex chromosomes and state:

   Chromosome configuration: _____ Sex of individual (M/F): ___ Syndrome: _____





PHOTOS: Cytogenetics Department, Waikato Hospital

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Examples of Aneuploidy in Human Sex Chromosomes

Sex chromosomes and chromosome condition	Apparent sex	Phenotype
XO, monosomic	Female	Turner syndrome
XX, disomic	Female	**Normal female**
XXX, trisomic	Female	Metafemale. Most appear normal; they have a greater tendency to criminality
XXXX, tetrasomic	Female	Rather like Down syndrome, low fertility and intelligence
XY, disomic	Male	**Normal male**
XYY, trisomic	Male	Jacob syndrome, apparently normal male, tall, aggressive
XXY, trisomic	Male	Klinefelter syndrome (infertile). Incidence rate 1 in 1000 live male births, with a maternal age effect.
XXXY, tetrasomic	Male	Extreme Klinefelter, mentally retarded

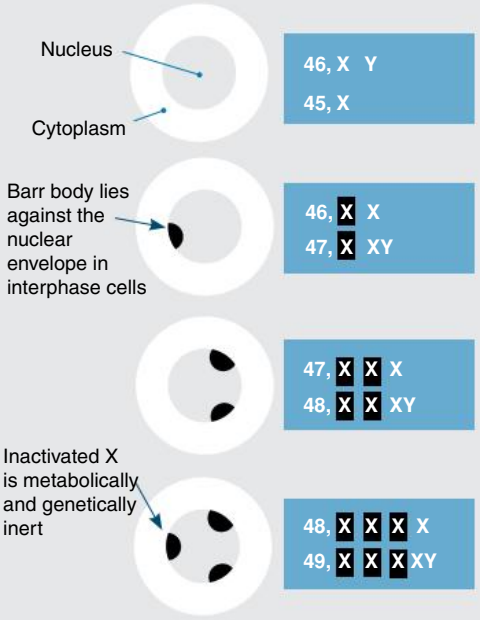
ABOVE: Features of selected aneuploidies in humans. Note that this list represents only a small sample of the possible sex chromosome aneuploidies in humans.

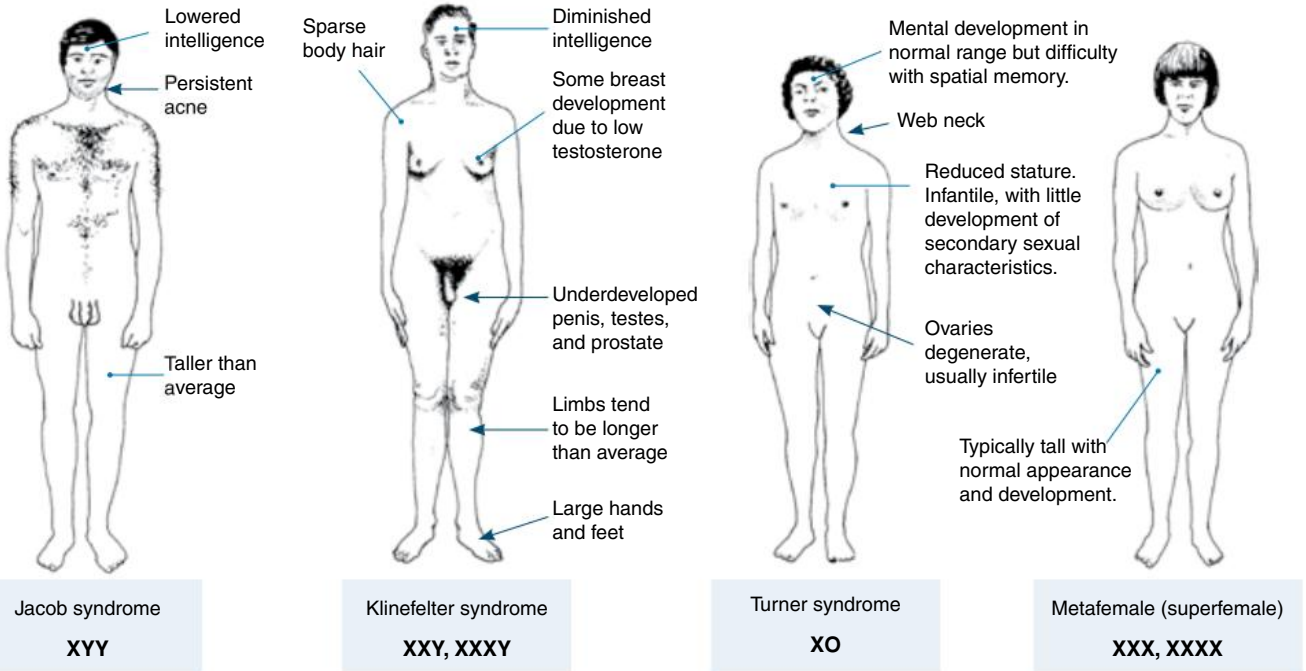
RIGHT: Symbolic representation of Barr body occurrence in various human karyotypes. The chromosome number is given first, and the inactive X chromosomes (Xi) are framed by a black box. Note that in aneuploid syndromes, such as those described here, all but one of the X chromosomes are inactivated, regardless of the number present.

If extra copies of X are inactivated, why do extra copies still produce the aneuploidy syndromes? This is because some of the genes on the Xi escape inactivation so the dosage of these non-silenced genes will differ as they escape inactivation.

Barr Bodies

In the nucleus of any non-dividing somatic cell, one of the X chromosomes condenses to form a visible piece of chromatin, called a **Barr body**. This chromosome is inactivated (Xi), so that only one X chromosome in a cell ever has its genes expressed. The inactivation is random, so Xi may be either the maternal homologue (from the mother) or the paternal homologue (from the father).





Jacob syndrome	Klinefelter syndrome	Turner syndrome	Metafemale (superfemale)
XYY	**XXY, XXXY**	**XO**	**XXX, XXXX**

5. State how many Barr bodies are present in each somatic cell for each of the following syndromes:

   (a) Jacob syndrome: _____ (b) Klinefelter syndrome: _____ (c) Turner syndrome: _____

6. Explain the consequence of X-chromosome inactivation in terms of the proteins encoded by the X chromosome genes:

   _____

   _____

7. State how many chromosomes for each set of homologues are present for the following forms of aneuploidy:

   (a) Nullisomy: _____ (c) Trisomy: _____

   (b) Monosomy: _____ (d) Polysomy: _____

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179 Mendel's Pea Plant Experiments

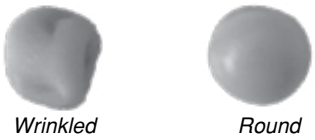
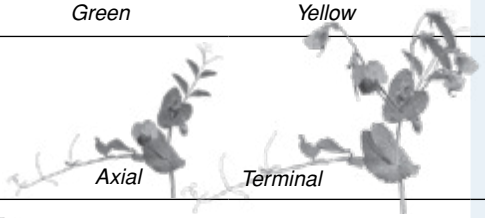
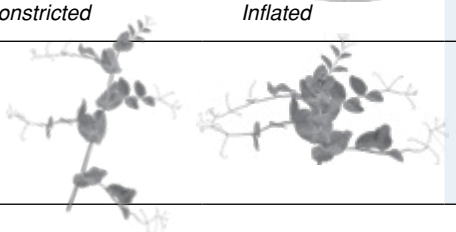
Key Idea: Many genes produce phenotypic traits that are inherited in predictable ratios, as shown by Mendel's pea experiments.



Gregor Mendel (1822-84), right, was an Austrian monk who carried out the pioneering studies of inheritance. Mendel bred pea plants to study the inheritance patterns of a number of **traits** (specific characteristics). He showed that characters could be masked in one generation but could reappear in later generations and proposed that inheritance involved the transmission of discrete units of inheritance from one generation to the next. At the time the mechanism of inheritance was unknown, but further research has provided an accepted mechanism and we know these units of inheritance are **genes**. The entire genetic makeup of an organism is its **genotype**.

Mendel examined six traits and found that they were inherited in predictable ratios, depending on the **phenotypes** (the physical appearance) of the parents. Some of his results from crossing heterozygous plants are tabulated below. The numbers in the results column represent how many offspring had those traits.

1. Study the results for each of the six experiments below. Determine which of the two phenotypes is dominant, and which is the recessive. Place your answers in the spaces in the **dominance** column in the table below.
2. Calculate the ratio of dominant phenotypes to recessive phenotypes (to two decimal places). The first one has been done for you (5474 ÷ 1850 = 2.96). Place your answers in the spaces provided in the table below:

Trait	Possible Phenotypes	Results	Dominance	Ratio
Seed shape	 *Wrinkled* *Round*	Wrinkled 1850 Round 5474 **TOTAL 7324**	Dominant: Round Recessive: Wrinkled	2.96 : 1
Seed colour	 *Green* *Yellow*	Green 2001 Yellow 6022 **TOTAL 8023**	Dominant: Recessive	
Pod colour	 *Green* *Yellow*	Green 428 Yellow 152 **TOTAL 580**	Dominant: Recessive	
Flower position	 *Axial* *Terminal*	Axial 651 Terminal 207 **TOTAL 858**	Dominant: Recessive	
Pod shape	 *Constricted* *Inflated*	Constricted 299 Inflated 882 **TOTAL 1181**	Dominant: Recessive	
Stem length		Tall 787 Dwarf 277 **TOTAL 1064**	Dominant: Recessive	

3. Mendel's experiments identified that two heterozygous parents should produce offspring in the ratio of three times as many dominant offspring to those showing the recessive phenotype.

   (a) Which three of Mendel's experiments provided ratios closest to the theoretical 3:1 ratio?

   _____

   (b) Suggest why these results deviated less from the theoretical ratio than the others: _____

   _____

   _____

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180 Mendel's Laws of Inheritance

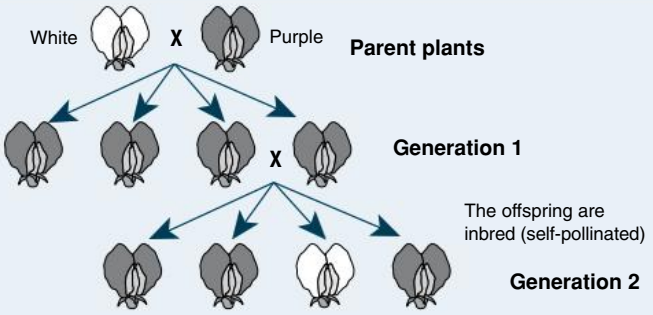
Key Idea: Genetic information is inherited from parents in discrete units called genes.

Mendel's laws of inheritance were based on his observations and govern how genes are passed to the offspring.

Particulate Inheritance

Characteristics of both parents are passed on to the next generation as discrete entities (genes).

This model explained many observations that could not be explained by the idea of blending inheritance, which was universally accepted prior to this theory. The trait for flower colour (right) appears to take on the appearance of only one parent plant in the first generation, but reappears in later generations.

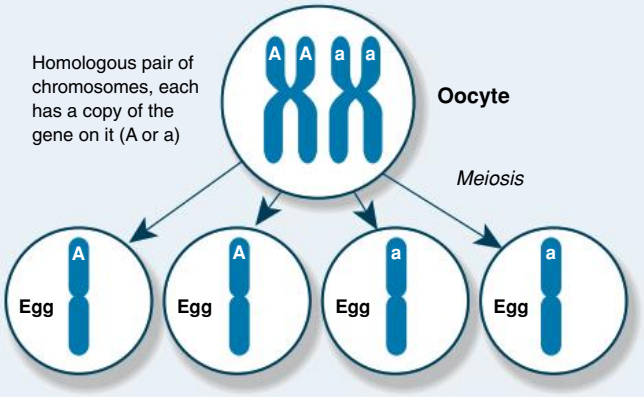


Law of Segregation

During gametic meiosis, the two members of any pair of alleles segregate unchanged and are passed into different gametes.

These gametes are eggs (ova) and sperm cells. The allele in the gamete will be passed on to the offspring.

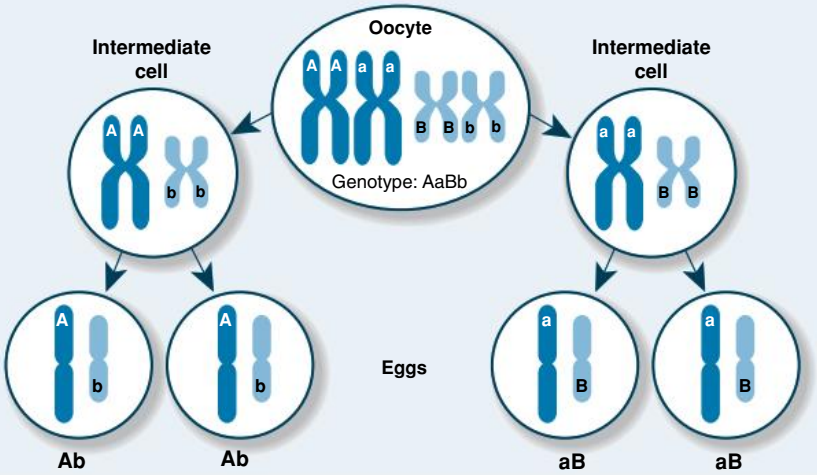
NOTE: This diagram has been simplified, omitting the stage where the second chromatid is produced for each chromosome.



Law of Independent Assortment

Allele pairs separate independently during gamete formation, and traits are passed on to offspring independently of one another (this is only true for unlinked genes).

This diagram shows are two genes (A and B) that code for different traits. Each of these genes is represented twice, one copy (allele) on each of two homologous chromosomes. The genes A and B are located on different chromosomes and, because of this, they will be inherited independently of each other i.e. the gametes may contain any combination of the parental alleles.



1. State the **property of genetic inheritance** that allows parent pea plants of different flower colour to give rise to flowers of a single colour in the first generation, with both parental flower colours reappearing in the following generation:

_____

2. The oocyte is the egg producing cell in the ovary of an animal. In the diagram illustrating the **law of segregation** above:

   (a) State the genotype for the oocyte (adult organism): _____

   (b) State the genotype of each of the **four** gametes: _____

   (c) State how many different kinds of gamete can be produced by this oocyte: _____

3. The diagram illustrating the **law of independent assortment** (above) shows only one possible result of the random sorting of the chromosomes to produce: Ab and aB in the gametes.
   (a) List another possible combination of genes (on the chromosomes) ending up in gametes from the same oocyte:

   _____

   (b) How many different gene combinations are possible for the oocyte? _____

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181 Describing Alleles

Key Idea: Eukaryotes generally have paired chromosomes. Each chromosome contains many genes and each gene may have a number of versions called alleles.

Sexually reproducing organisms usually have paired sets of chromosomes, one set from each parent. The equivalent chromosomes that form a pair are termed **homologues**. They carry equivalent sets of genes, but there is the potential for different versions of a gene (**alleles**) to exist in a population.

Homologous Chromosomes

In sexually reproducing organisms, most cells have a homologous pair of chromosomes (one coming from each parent). This diagram shows the position of three different genes on the same chromosome that control three different traits (A, B and C).

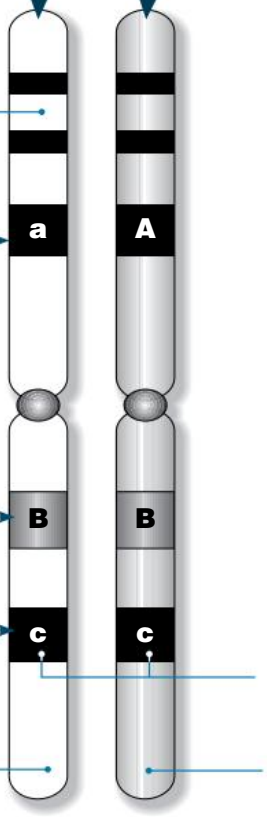
Chromosomes are formed from DNA and proteins. DNA tightly winds around special proteins to form the chromosome.

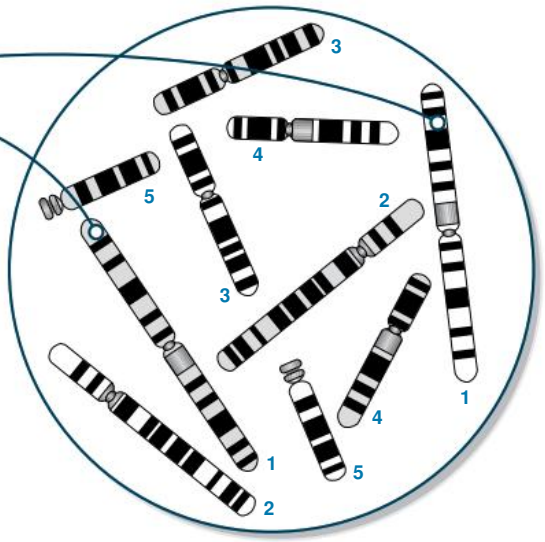
Having two different versions (alleles) of gene A is a **heterozygous** condition. Only the dominant allele (A) will be expressed. Alleles differ by only a few bases.

When both chromosomes have identical copies of the dominant allele for gene B the organism is **homozygous dominant** for that gene.

When both chromosomes have identical copies of the recessive allele for gene C the organism is said to be **homozygous recessive** for that gene.

Maternal chromosome originating from the egg of this individual's mother.





This diagram shows the complete chromosome complement for a hypothetical organism. It has a total of ten chromosomes, as five, nearly identical pairs (each pair is numbered). Each parent contributes one chromosome to the pair. The pairs are called **homologues** or **homologous pairs**. Each homologue carries an identical assortment of genes, but the version of the gene (the allele) from each parent may differ.

A gene is the unit of heredity. Genes occupying the same **locus** or position on a chromosome code for the same trait (e.g. dimpled chin).

Paternal chromosome originating from the sperm of this individual's father.

1. Define the following terms used to describe the allele combinations in the genotype for a given gene:

   (a) Heterozygous: _____

   (b) Homozygous dominant: _____

   (c) Homozygous recessive: _____

2. For a gene given the symbol '**A**', name the alleles present in an organism that is identified as:

   (a) Heterozygous: _____ (b) Homozygous dominant: _____ (c) Homozygous recessive: _____

3. What is a **homologous pair** of chromosomes? _____

4. Discuss the significance of genes existing as **alleles**: _____

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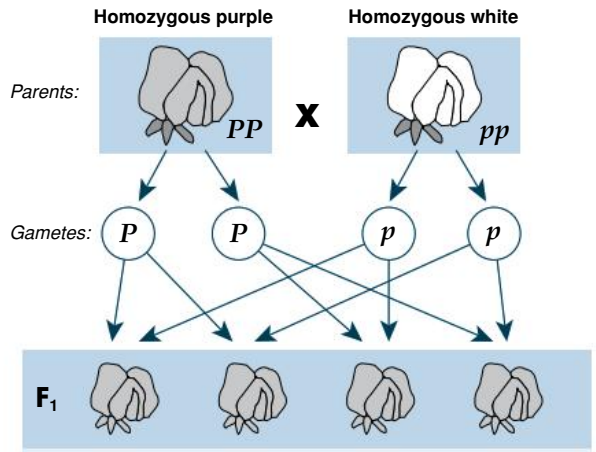
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182 Basic Genetic Crosses

Key Idea: The outcome of a cross depends on the parental genotypes and can be determined using Punnett squares.

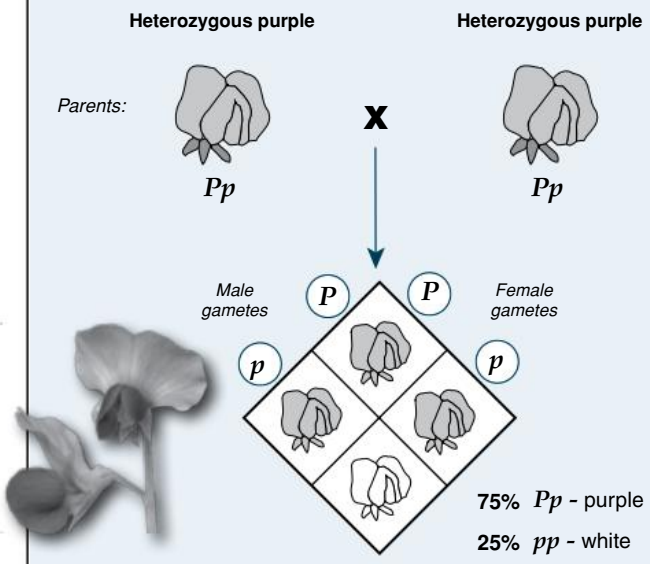
Examine the diagrams below on monohybrid (one gene) and dihybrid (two gene) inheritance. The F_1 generation describes the offspring of a cross between **true-breeding** (homozygous) parents. A **back cross** is a cross between an offspring and one of its parents. If the back cross is to a homozygous recessive, it can be used as a test cross (see opposite).

Monohybrid Cross F_1



A true-breeding organism is homozygous for the gene involved. The F_1 offspring of a cross between two **true breeding** parent plants are all purple (*Pp*).

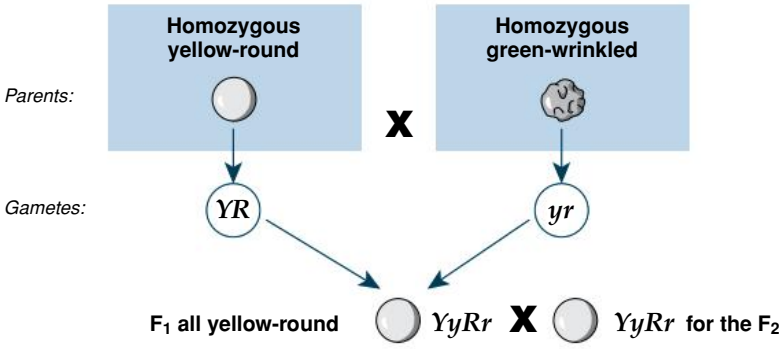
Monohybrid Cross F_2



A cross between the F_1 offspring (*Pp x Pp*) would yield a 3:1 ratio in the F_2 of purple (*PP, Pp, Pp*) to white (*pp*).

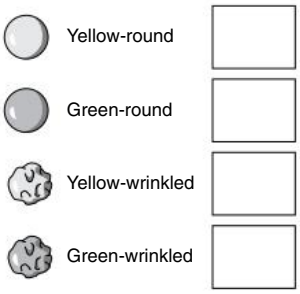
Dihybrid Cross

A dihybrid cross studies the inheritance patterns of two genes. In pea seeds, yellow color (*Y*) is dominant to green (*y*) and round shape (*R*) is dominant to wrinkled (r). Each **true breeding** parental plant has matching alleles for each of these characters (*YYRR* or *yyrr*). F_1 offspring will all have the same genotype and phenotype (yellow-round: *YyRr*).



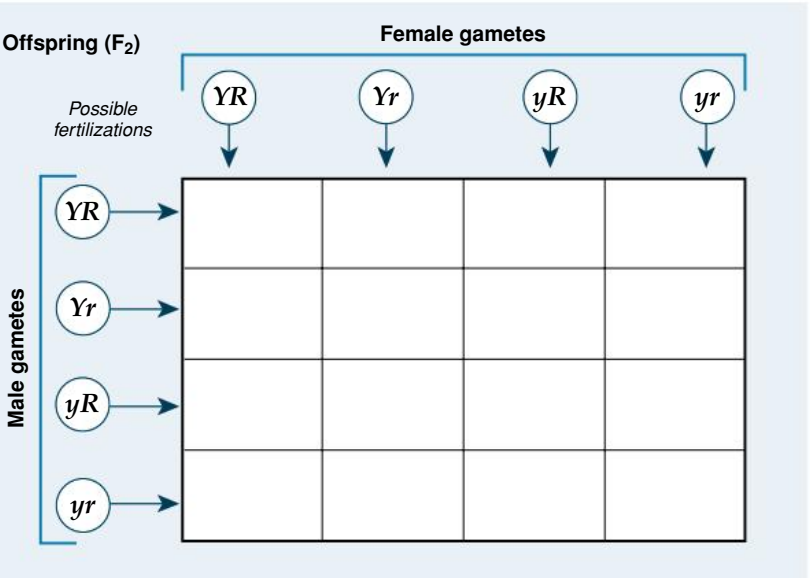
1. Fill in the Punnett square (below right) to show the genotypes of the F_2 generation.

2. In the boxes below, use fractions to indicate the numbers of each phenotype produced from this cross.



3. Express these numbers as a ratio:

_____



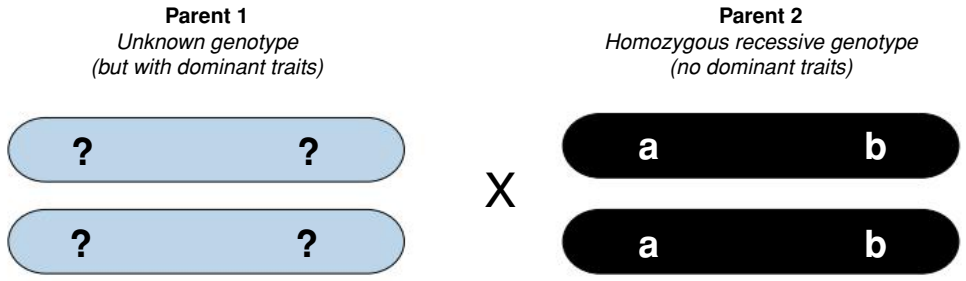
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183 The Test Cross

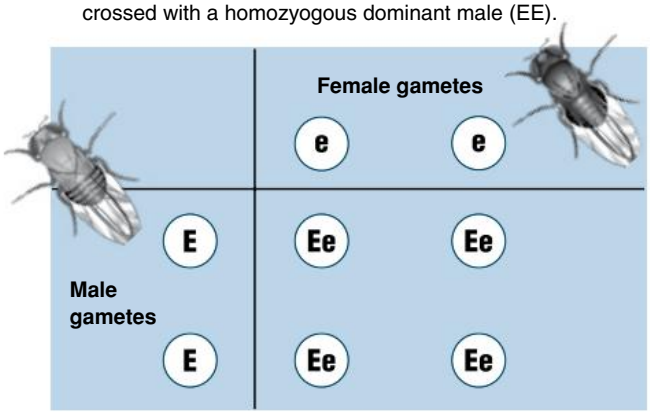
Key Idea: If an individual's genotype is unknown it can be determined using a test cross.

It is not always possible to determine an organism's genotype by its appearance because gene expression is complicated by patterns of dominance and by gene interactions. The **test cross** was developed by Mendel as a way to establish the genotype of an organism with the dominant phenotype for a particular trait. The principle is simple. The individual with the unknown genotype is bred with a homozygous recessive individual for the trait(s) of interest. The homozygous recessive can produce only one type of allele (recessive), so the phenotypes of the offspring will reveal the genotype of the unknown parent (below). The test cross can be used to determine the genotype of single genes or multiple genes.



The common fruit fly (*Drosophila melanogaster*) is often used to illustrate basic principles of inheritance because it has several genetic markers whose phenotypes are easily identified. Once such phenotype is body colour. Wild type (normal) *Drosophila* have yellow-brown bodies. The allele for yellow-brown body colour (E) is dominant. The allele for an ebony coloured body (e) is recessive. The test crosses below show the possible outcomes for an individual with homozygous and heterozygous alleles for ebony body colour.

A. A homozygous recessive female (ee) with an ebony body is crossed with a homozyogous dominant male (EE).

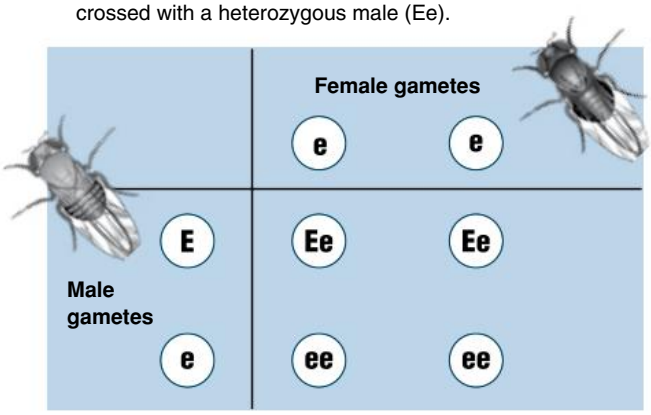


Cross A:

(a) Genotype frequency: 100% Ee

(b) Phenotype frequency: 100% yellow-brown

B. A homozygous recessive female (ee) with an ebony body is crossed with a heterozygous male (Ee).



Cross B:

(a) Genotype frequency: 50% Ee, 50% ee

(b) Phenotype frequency: 50% yellow-brown, 50% ebony

1. In *Drosophila*, the allele for brown eyes (b) is recessive, while the red eye allele (B) is dominant. How would you set up a **two gene test cross** to determine the genotype of a male who has a normal body colour and red eyes?

2. List all of the **possible genotypes** for the male *Drosophila*: _____

3. 50% of the resulting progeny are yellow-brown bodies with red eyes, and 50% have ebony bodies with red eyes.

   (a) What is the genotype of the male *Drosophila*? _____

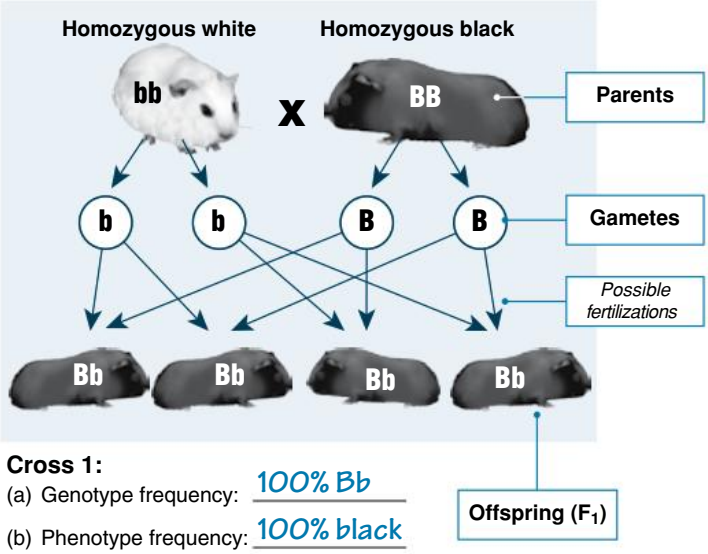
   (b) Explain your answer: _____

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184 Monohybrid Cross

Key Idea: A monohybrid cross studies the inheritance pattern of one gene. The offspring of these crosses occur in predictable ratios.

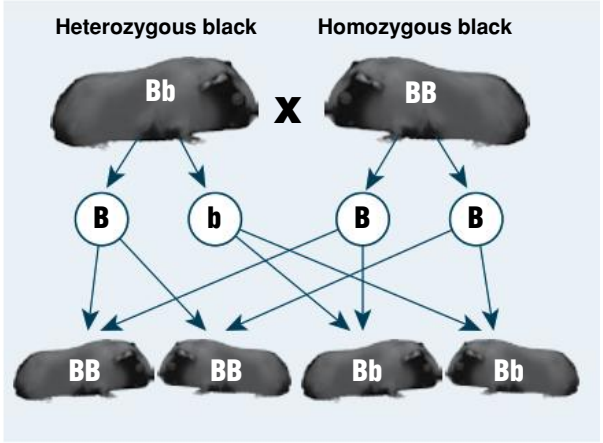
In this activity, you will examine six types of matings possible for a pair of alleles governing coat colour in guinea pigs. A dominant allele (**B**) produces **black** hair and its recessive allele (**b**), produces white. Each parent can produce two types of gamete by meiosis. Determine the **genotype** and **phenotype frequencies** for the crosses below. For crosses 3 to 6, also determine the gametes produced by each parent (write these in the circles) and offspring genotypes and phenotypes (write these inside the offspring shapes).



Cross 1:

(a) Genotype frequency: 100% Bb

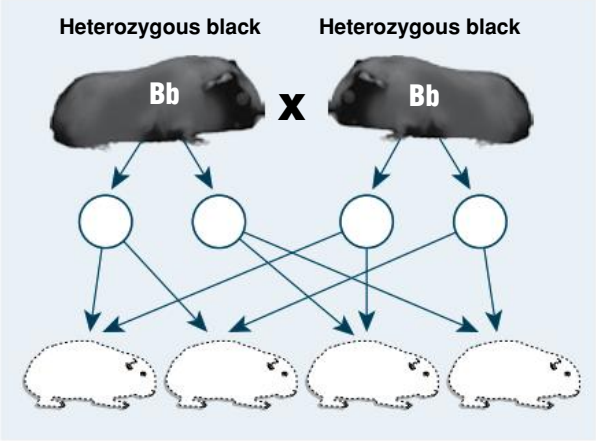
(b) Phenotype frequency: 100% black



Cross 2:

(a) Genotype frequency: _____

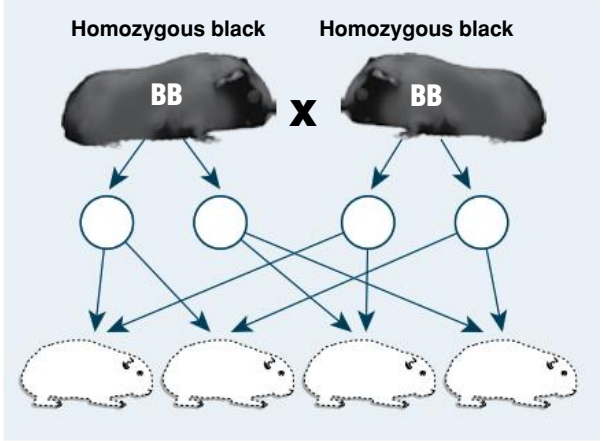
(b) Phenotype frequency: _____



Cross 3:

(a) Genotype frequency: _____

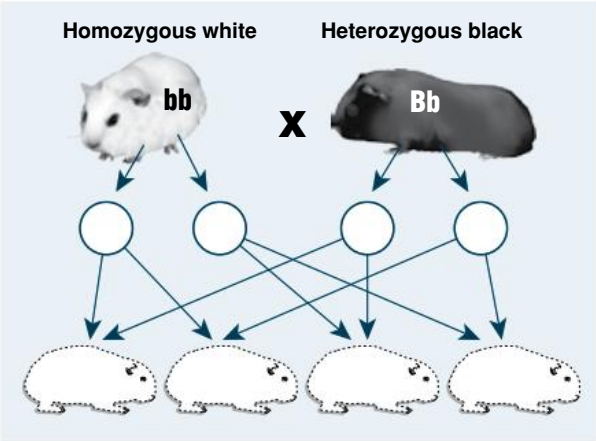
(b) Phenotype frequency: _____



Cross 4:

(a) Genotype frequency: _____

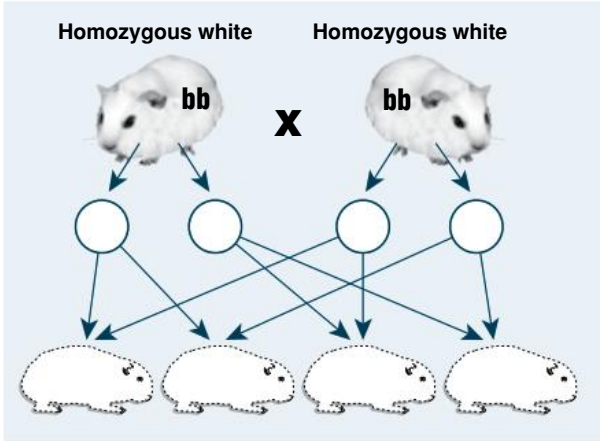
(b) Phenotype frequency: _____



Cross 5:

(a) Genotype frequency: _____

(b) Phenotype frequency: _____



Cross 6:

(a) Genotype frequency: _____

(b) Phenotype frequency: _____

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185 Problems Involving Monohybrid Inheritance

Key Idea: For monohybrid crosses involving autosomal unlinked genes, the offspring appear in predictable ratios.

The following problems involve Mendelian crosses. The alleles involved are associated with various phenotypic traits controlled by a single gene. The problems are to give you practice in problem solving using Mendelian genetics.



1. A dominant gene (**W**) produces wire-haired texture in dogs; its recessive allele (**w**) produces smooth hair. A group of heterozygous wire-haired individuals are crossed and their F_1 progeny are then test-crossed. Determine the expected genotypic and phenotypic ratios among the **test cross** progeny:

2. In sheep, black wool is due to a recessive allele (**b**) and white wool to its dominant allele (**B**). A white ram is crossed to a white ewe. Both animals carry the black allele (b). They produce a white ram lamb, which is then back crossed to the female parent. Determine the probability of the **back cross** offspring being black:



3. A recessive allele, a, is responsible for albinism, an inability to produce or deposit melanin in tissues. Humans and a variety of other animals can exhibit this phenotype. In each of the following cases, determine the possible genotypes of the mother and father, and of their children:

   (a) Both parents have normal phenotypes; some of their children are albino and others are unaffected:

   (b) Both parents are albino and have only albino children:

   (c) The woman is unaffected, the man is albino, and they have one albino child and three unaffected children:

4. Chickens with shortened wings and legs are called creepers. When creepers are mated to normal birds, they produce creepers and normals with equal frequency. When creepers are mated to creepers they produce two creepers to one normal. Crosses between normal birds produce only normal progeny. Explain these results:

5. In a dispute over parentage, the mother of a child with blood group O identifies a male with blood group A as the father. The mother is blood group B. Draw Punnett squares to show possible genotype/phenotype outcomes to determine if the male is the father and the reasons (if any) for further dispute:

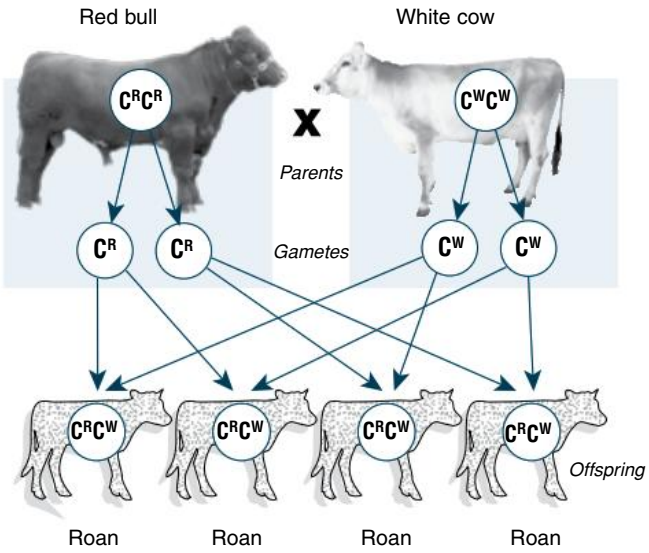
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LINK 188 LINK 187 LINK 184 TEST

186 Codominance

Key Idea: In codominance, neither allele is recessive and both alleles are equally and independently expressed in the heterozygote.

Codominance is an inheritance pattern in which both alleles in a heterozygote contribute to the phenotype and both alleles are **independently** and **equally expressed**. Examples include the human blood group AB and certain coat colours in horses and cattle. Reddish coat colour is equally dominant with white. Animals that have both alleles have coats that are roan (both red and white hairs are present).




A roan shorthorn heifer

In the shorthorn cattle breed, coat colour is inherited. White shorthorn parents always produce calves with white coats. Red parents always produce red calves. However, when a red parent mates with a white one, the calves have a coat colour that is different from either parent; a mixture of red and white hairs, called roan. Use the example (left) to help you to solve the problems below.

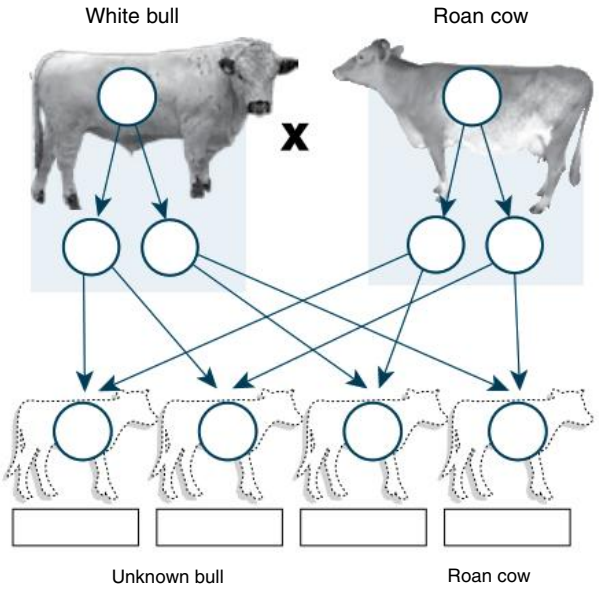
1. Explain how codominance of alleles can result in offspring with a phenotype that is different from either parent:

2. A white bull is mated with a roan cow (right):

   (a) Fill in the spaces to show the genotypes and phenotypes for parents and calves:

   (b) What is the phenotype ratio for this cross?

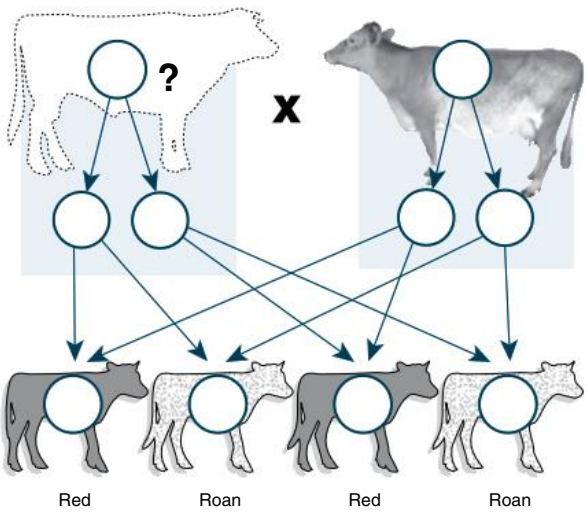
   (c) How could a cattle farmer control the breeding so that the herd ultimately consisted of only red cattle:



3. A farmer has only roan cattle on his farm. He suspects that one of the neighbours' bulls may have jumped the fence to mate with his cows earlier in the year because half the calves born were red and half were roan. One neighbour has a red bull, the other has a roan.

   (a) Fill in the spaces (right) to show the genotype and phenotype for parents and calves.

   (b) Which bull serviced the cows? **red** or **roan** (*delete one*)



4. Describe the classical phenotypic ratio for a codominant gene resulting from the cross of two heterozygous parents (e.g. a cross between two roan cattle):

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187 Codominance in Multiple Allele Systems

Key Idea: The human ABO blood group system is a multiple allele system involving the codominant alleles *A* and *B* and the recessive allele *O*.

The four common blood groups of the human 'ABO blood group system' are determined by three alleles: ***A***, ***B***, and ***O***. The ABO antigens consist of sugars attached to the surface of red blood cells. The alleles code for enzymes (proteins) that join these sugars together. The allele ***O*** produces a non-functioning enzyme that is unable to make any changes to the basic antigen (sugar) molecule. The other two alleles (***A***, ***B***) are **codominant** and are expressed equally. They each produce a different functional enzyme that adds a different, specific sugar to the basic sugar molecule. The blood group A and B antigens are able to react with antibodies present in the blood of other people so blood must always be matched for transfusion.

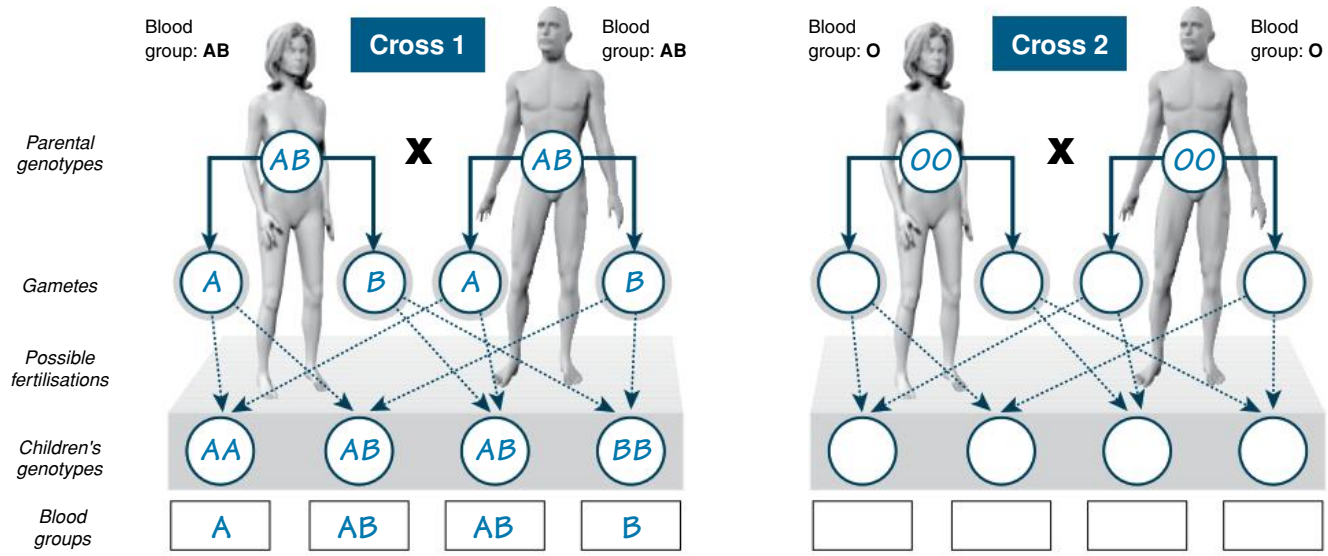
Recessive allele: ***O*** produces a non-functioning protein
Dominant allele: ***A*** produces an enzyme which forms **A antigen**
Dominant allele: ***B*** produces an enzyme which forms **B antigen**

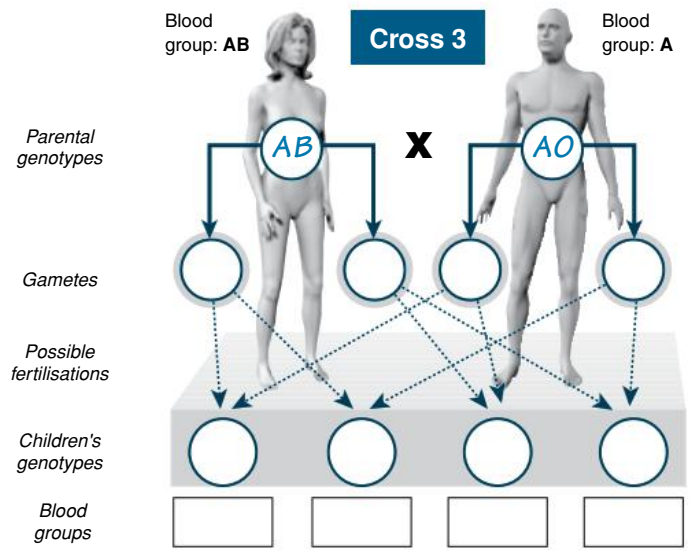
If a person has the ***AO*** allele combination then their blood group will be group **A**. The presence of the recessive allele has no effect on the blood group in the presence of a dominant allele. Another possible allele combination that can create the same blood group is ***AA***.

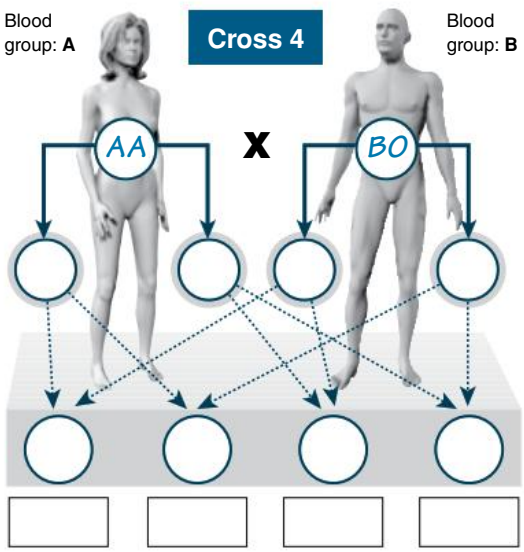
Blood group (phenotype)	Possible genotypes	Frequency in Australia
O	*OO*	49%
A	*AA AO*	38%
B		10%
AB		3%

1. Use the information above to complete the table for the possible genotypes for blood group B and group AB.

2. Below are four crosses possible between couples of various blood group types. The first example has been completed for you. Complete the genotype and phenotype for the other three crosses below:







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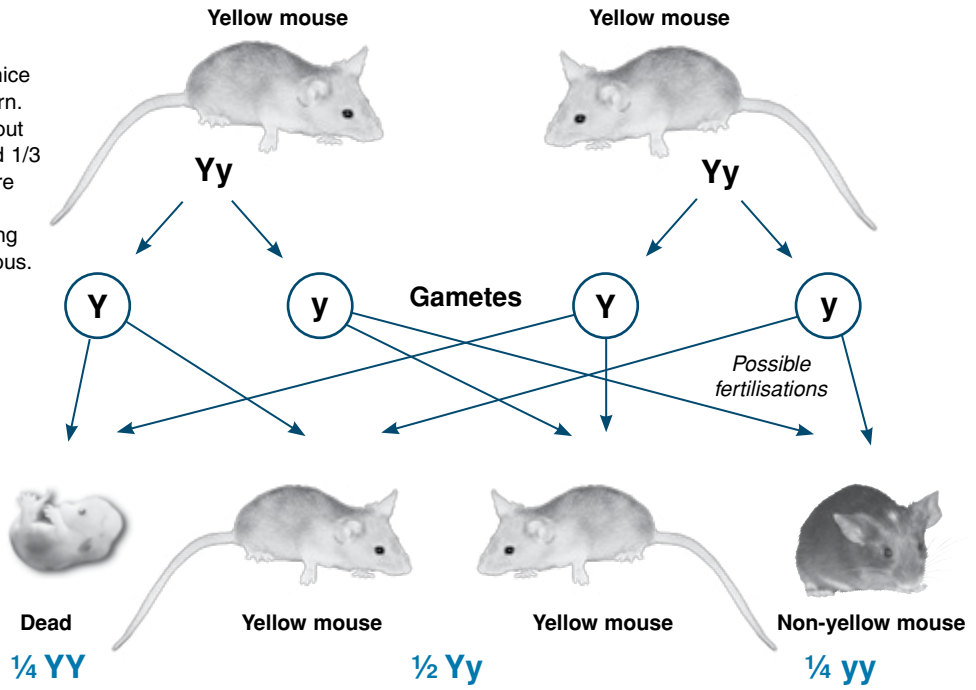
LINK 186 LINK 93 WEB 187 KNOW

188 Lethal Alleles

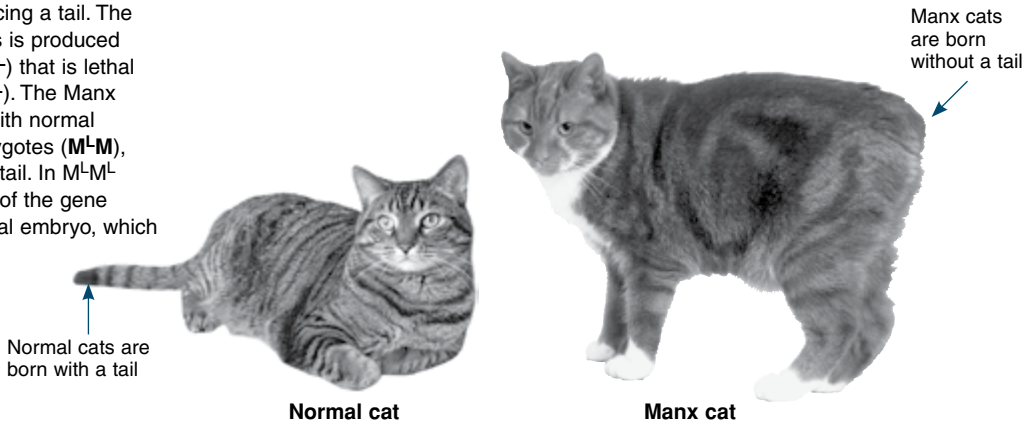
Key Idea: Lethal alleles are gene mutations resulting in a non-functional gene product which may affect survival.

Some lethal alleles are fully dominant and kill in one dose in the heterozygote. Others, such as in the **Manx** cat and yellow mice (below), produce viable offspring with a recognisable phenotype in the heterozygote. In some lethal alleles, the lethality is fully recessive and the alleles produce no detectable effect in the heterozygote at all. Furthermore, lethal alleles may take effect at different stages in development (e.g. in juveniles or, as in **Huntington's disease**, in adults).

When **Lucien Cuenot** investigated inheritance of coat colour in yellow mice in 1905, he reported a peculiar pattern. When he mated two yellow mice, about 2/3 of their offspring were yellow, and 1/3 were non-yellow. This was a departure from the expected Mendelian ratio of 3:1. A test cross of the yellow offspring showed that they were all heterozygous.



Cats possess a gene for producing a tail. The tailless **Manx** phenotype in cats is produced by the presence of an allele ($\mathbf{M^L}$) that is lethal in the homozygous state ($\mathbf{M^LM^L}$). The Manx allele (M^L) severely interferes with normal spinal development. In heterozygotes ($\mathbf{M^LM}$), this results in the absence of a tail. In M^LM^L homozygotes, the double dose of the gene produces an extremely abnormal embryo, which does not survive.



1. Distinguish between recessive lethal alleles and dominant lethal alleles: _____

_____

_____

2. In Manx cats, the allele for taillessness ($\mathbf{M^L}$) is incompletely dominant over the recessive allele for normal tail ($\mathbf{M}$). Tailless Manx cats are heterozygous ($\mathbf{M^LM}$) and carry a recessive allele for normal tail. Normal tailed cats are $\mathbf{MM}$. A cross between two Manx (tailless) cats, produces two Manx to every one normal tailed cat (not a regular 3 to 1 Mendelian ratio).

   (a) State the genotypes arising from this type of cross: _____

   (b) State the phenotype ratio of Manx to normal cats and explain why it is not the expected 3:1 ratio: _____

   _____

3. Explain why Huntington disease persists in the human population when it is caused by a lethal, dominant allele:

_____

_____

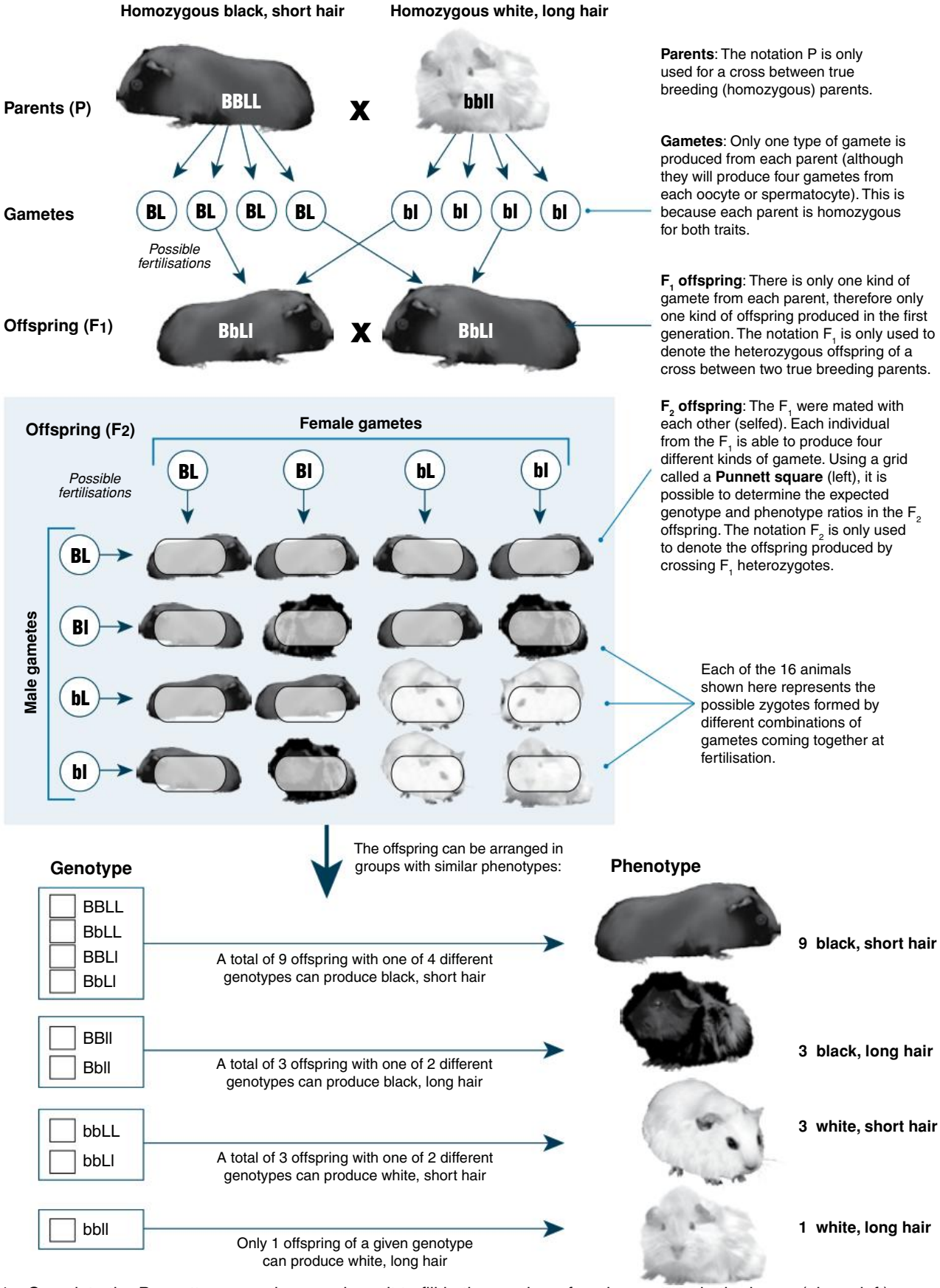
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189 Dihybrid Cross

Key Idea: A dihybrid cross studies the inheritance pattern of two genes. In crosses involving unlinked autosomal genes, the offspring occur in predictable ratios.

There are four types of gamete produced in a cross involving two genes, where the genes are carried on separate chromosomes and are sorted independently of each other during meiosis. The two genes in the example below are on separate chromosomes and control two unrelated characteristics, **hair colour** and **coat length**. Black (B) and short (L) are dominant to white and long.



1. Complete the Punnett square above and use it to fill in the number of each genotype in the boxes (above left).

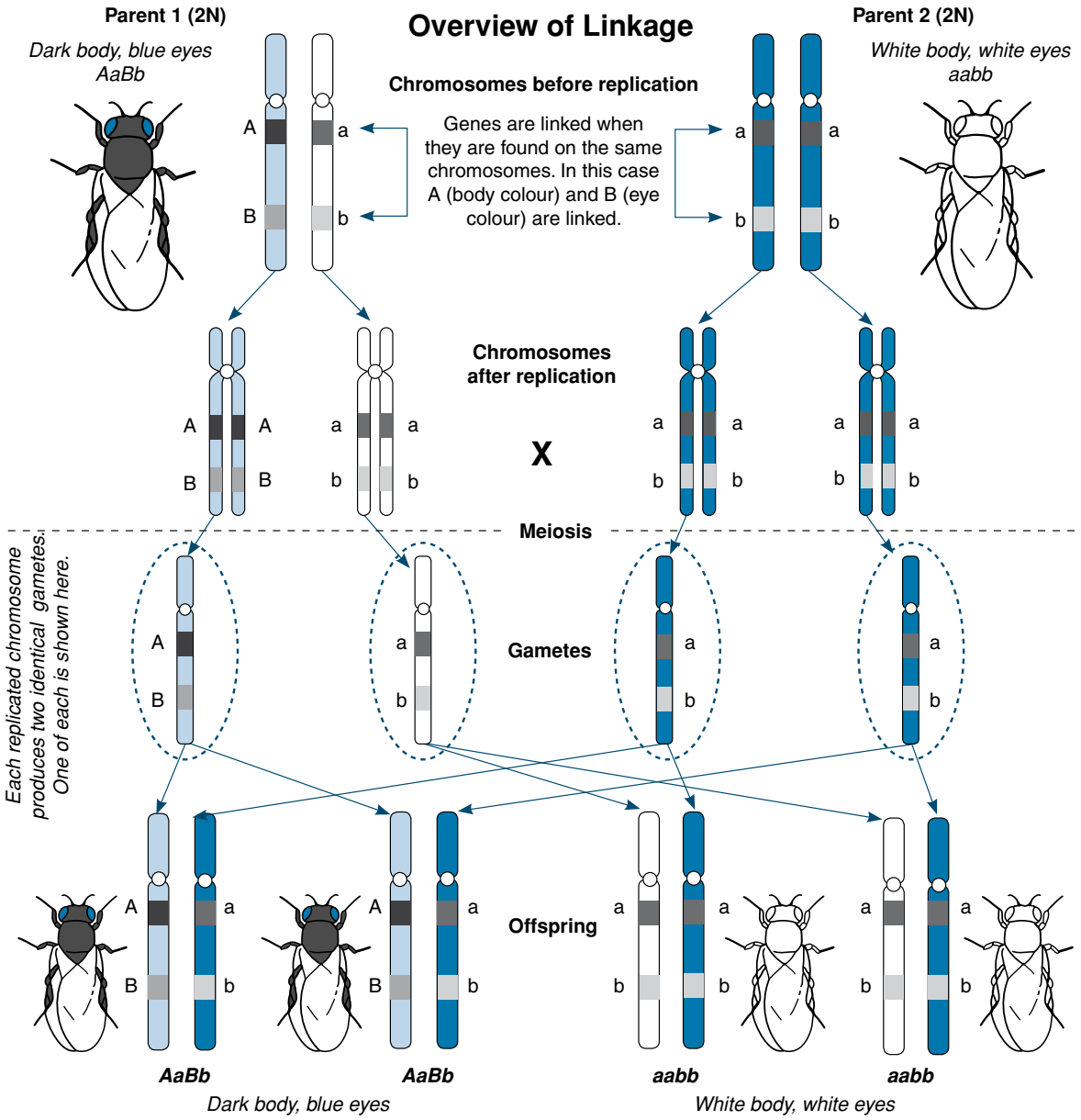
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LINK 192 LINK 191 LINK 182 WEB 189 KNOW

190 Inheritance of Linked Genes

Key Idea: Linked genes are genes found on the same chromosome and tend to be inherited together. Linkage reduces the genetic variation in the offspring.

Genes are **linked** when they are on the same chromosome. Linked genes tend to be inherited together and the extent of crossing over depends on how close together they are on the chromosome. In genetic crosses, linkage is indicated when a greater proportion of the offspring from a cross are of the parental type (than would be expected if the alleles were on separate chromosomes and assorting independently). Linkage reduces the genetic variation that can be produced in the offspring.



Possible Offspring

Only two kinds of genotype combinations are possible. They are they same as the parent genotype.

1. What is the effect of **linkage** on the inheritance of genes? _____

_____

_____

2. Explain how linkage decreases the amount of genetic variation in the offspring: _____

_____

_____

_____

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An Example of Linked Genes in *Drosophila*

The genes for wing shape and body colour are linked (they are on the same chromosome).

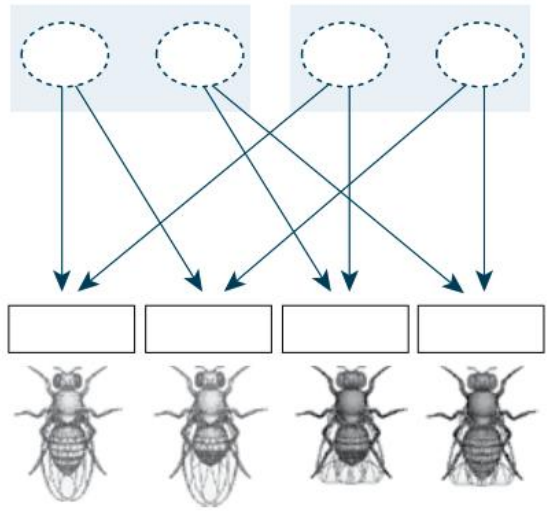




	Wild type female	Mutant male
Parent		
Phenotype	Straight wing Grey body	Curled wing Ebony body
Genotype	Cucu Ebeb	cucu ebeb
Linkage	*Cu Eb* *cu eb*	*cu eb* *cu eb*

Meiosis

Gametes from female fly (N) ***Gametes from male fly (N)***



Sex of offspring is irrelevant in this case

Contact **Newbyte Educational Software** for details of their superb *Drosophila Genetics* software package which includes coverage of linkage and recombination. *Drosophila* images © Newbyte Educational Software.

Drosophila and Linked Genes

In the example shown left, wild type alleles are dominant and are given an upper case symbol of the mutant phenotype (Cu or Eb). This notation used for *Drosophila* departs from the convention of using the dominant gene to provide the symbol. This is necessary because there are many mutant alternative phenotypes to the wild type (e.g. curled and vestigial wings). A lower case symbol of the wild type (e.g. ss for straight wing) would not indicate the mutant phenotype involved.

Drosophila melanogaster is known as a model organism. Model organisms are used to study particular biological phenomena, such as mutation. *Drosophila melanogaster* is particularly useful because it produces such a wide range of heritable mutations. Its short reproduction cycle, high offspring production, and low maintenance make it ideal for studying in the lab.



Drosophila melanogaster examples showing variations in eye and body colour. The wild type is marked with a w in the photo above.

3. Complete the linkage diagram above by adding the gametes in the ovals and offspring genotypes in the rectangles.

4. (a) List the possible genotypes in the offspring (above, left) if genes Cu and Eb had been on **separate chromosomes**:

_____

(b) If the female *Drosophila* had been homozygous for the dominant wild type alleles (CuCu EbEb), state:

The genotype(s) of the F_1: _____ The phenotype(s) of the F_1: _____

5. A second pair of *Drosophila* are mated. The female genotype is Vgvg EbEb (straight wings, grey body), while the male genotype is vgvg ebeb (vestigial wings, ebony body). Assuming the genes are linked, carry out the cross and list the genotypes and phenotypes of the offspring. Note vg = vestigial (no) wings:

_____

_____

_____

The genotype(s) of the F_1: _____ The phenotype(s) of the F_1: _____

6. Explain why *Drosophila* are often used as model organisms in the study of genetics: _____

_____

_____

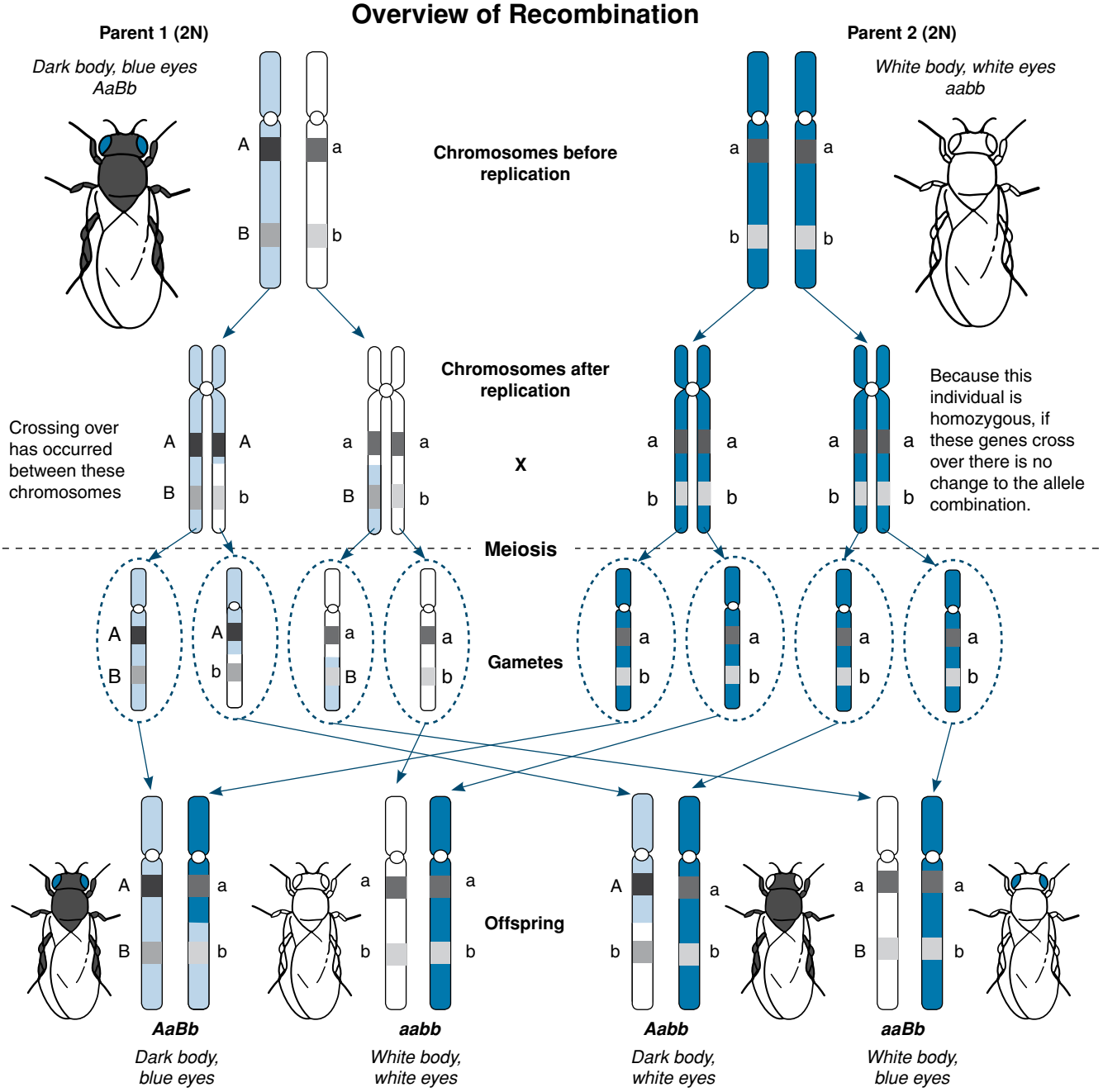
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191 Recombination and Dihybrid Inheritance

Key Idea: Recombination is the exchange of alleles between homologous chromosomes as a result of crossing over. Recombination increases the genetic variation in the offspring. The alleles of parental linkage groups separate and new associations of alleles are formed in the gametes. Offspring formed from these gametes are called **recombinants** and show combinations of characteristics not seen in the parents.

In contrast to linkage, recombination increases genetic variation in the offspring. Recombination between the alleles of parental linkage groups is indicated by the appearance of non-parental types in the offspring, although not in the numbers that would be expected had the alleles been on separate chromosomes (independent assortment).

Overview of Recombination



Non-recombinant offspring

These two offspring show allele combinations that are expected as a result of independent assortment during meiosis. Also called parental types.

Recombinant offspring

These two offspring show unexpected allele combinations. They can only arise if one of the parent's chromosomes has undergone crossing over.

1. Describe the effect of **recombination** on the inheritance of genes: _____

_____

_____

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An Example of Recombination

In the female parent, crossing over occurs between the linked genes for wing shape and body colour

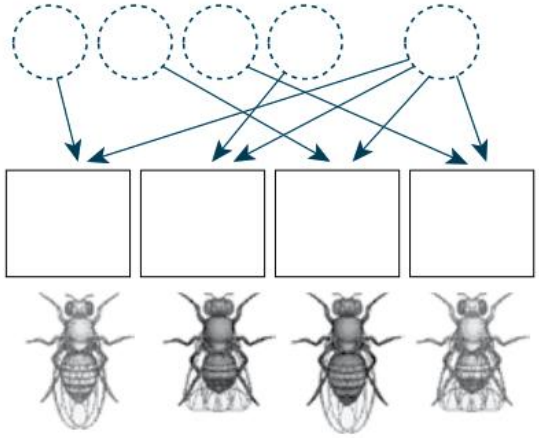




	Wild type female	Mutant male
Parent		
Phenotype	Straight wing Grey body	Curled wing Ebony body
Genotype	Cucu Ebeb	cucu ebeb
Linkage	*Cu* *Eb* *cu* *eb*	*cu* *eb* *cu* *eb*

Meiosis

Gametes from female fly (N)	***Gametes from male fly (N)***
Crossing over has occurred, giving four types of gametes	Only one type of gamete is produced in this case



Non-recombinant offspring	Recombinant offspring

The sex of the offspring is irrelevant in this case

Contact **Newbyte Educational Software** for details of their superb *Drosophila Genetics* software package which includes coverage of linkage and recombination. *Drosophila* images © Newbyte Educational Software.

The cross (left) uses the same genotypes as the previous activity but, in this case, crossing over occurs between the alleles in a linkage group in one parent. The symbology used is the same.

Recombination Produces Variation

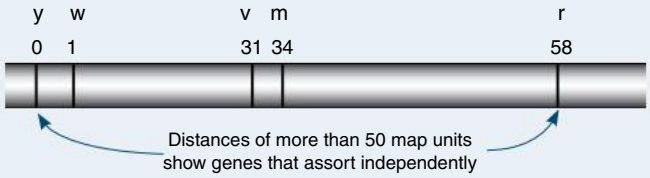
If crossing over does not occur, the possible combinations in the gametes is limited. **Crossing over and recombination increase the variation in the offspring**. In humans, even without crossing over, there are approximately $(2^{23})^2$ or 70 trillion genetically different zygotes that could form for every couple. Taking crossing over and recombination into account produces $(4^{23})^2$ or 5000 trillion trillion genetically different zygotes for every couple.



Family members may resemble each other, but they'll never be identical (except for identical twins).

Using Recombination

Analysing recombination gave geneticists a way to map the genes on a chromosome. Crossing over is less likely to occur between genes that are close together on a chromosome than between genes that are far apart. By counting the number of offspring of each phenotype, the **frequency of recombination** can be calculated. The higher the frequency of recombination between two genes, the further apart they must be on the chromosome.



Map of the X chromosome of *Drosophila*, showing the relative distances between five different genes (in map units).

2. Complete the recombination diagram above, adding the gametes in the ovals and offspring genotypes and phenotypes in the rectangles:

3. Explain how recombination increases the amount of genetic variation in offspring: _____

_____

_____

4. Explain why it is not possible to have a recombination frequency of greater than 50% (half recombinant progeny):

_____

_____

5. A second pair of *Drosophila* are mated. The female is Cucu YY (straight wing, grey body), while the male is Cucu yy (straight wing, yellow body). Assuming recombination, perform the cross and list the offspring genotypes and phenotypes:

_____

_____

_____

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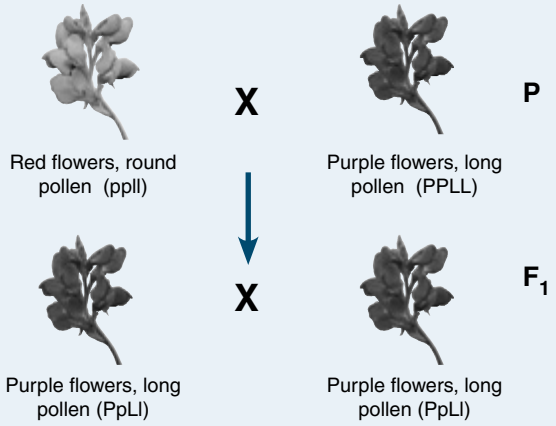
192 Detecting Linkage in Dihybrid Crosses

Key Idea: Linkage between genes can be detected by observing the phenotypic ratios in the offspring.

Shortly after the rediscovery of Mendel's work early in the 20th century, it became apparent that his ratios of 9:3:3:1 for heterozygous dihybrid crosses did not always hold true.

Experiments on sweet peas by William Bateson and Reginald Punnett, and on *Drosophila* by Thomas Hunt Morgan, showed that there appeared to be some kind of coupling between genes. This coupling, which we now know to be linkage, did not follow any genetic relationship known at the time.

Sweet Pea Cross



Bateson and Punnett studied sweet peas in which purple flowers (P) are dominant to red (p), and long pollen grains (L) are dominant to round (l). If these genes were unlinked, the outcome of an cross between two heterozygous sweet peas should have been a 9:3:3:1 ratio.

Table 1: Sweet Pea Cross Results

	Observed	Expected
Purple long (P_L_)	284	
Purple round (P_ll)	21	
Red long (ppL_)	21	
Red round (ppll)	55	
Total	381	381

1. Fill in the missing numbers in the **expected** column of **Table 1**, remembering that a 9:3:3:1 ratio is expected:

2. (a) Fill in the missing numbers in the **expected** column of **Table 2**, remembering that a 1:1:1:1 ratio is expected:

   (b) Add the gamete type (parental/recombinant) to the gamete type column in Table 2:

   (c) What type of cross did Morgan perform here?

   _____

3. (a) Use the pedigree chart below to determine if nail-patella syndrome is dominant or recessive, giving reasons for your choice:

   _____

   _____

   _____

   _____

   (b) What evidence is there that nail-patella syndrome is linked to the ABO blood group locus?

   _____

   _____

   _____

   (c) Suggest a likely reason why individual III-3 is not affected despite carrying the B allele:

   _____

   _____

   _____

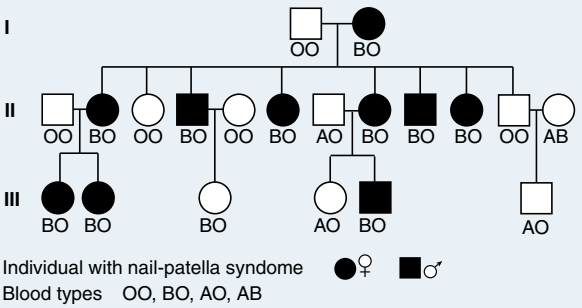
Morgan performed experiments to investigate linked genes in *Drosophila*. He crossed a heterozygous red-eyed normal-winged (Prpr Vgvg) fly with a homozygous purple-eyed vestigial-winged (prpr vgvg) fly. The table (below) shows the outcome of the cross.



Table 2: *Drosophila* Cross Results

Genotype	Observed	Expected	Gamete type
Prpr Vgvg	1339	710	Parental
prpr Vgvg	152		
Prpr vgvg	154		
prpr vgvg	1195		
Total	2840	2840	

Pedigree for Nail-Patella Syndrome



Linked genes can be detected by pedigree analysis. The diagram above shows the pedigree for the inheritance of nail-patella syndrome, which results in small, poorly developed nails and kneecaps in affected people. Other body parts such as elbows, chest, and hips can also be affected. The nail-patella syndrome gene is linked to the ABO blood group locus.

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193 Problems Involving Dihybrid Inheritance

Key Idea: For dihybrid crosses involving autosomal unlinked genes, the offspring appear in predictable ratios.

Test your understanding of dihybrid inheritance by solving problems involving the inheritance of two genes.

1. In cats, the following alleles are present for coat characteristics: black (B), brown (b), short (L), long (l), tabby (T), blotched tabby (tb). Use the information to complete the dihybrid crosses below:



(a) A black short haired (**BBLl**) male is crossed with a black long haired (**Bbll**) female. Determine the genotypic and phenotypic ratios of the offspring:

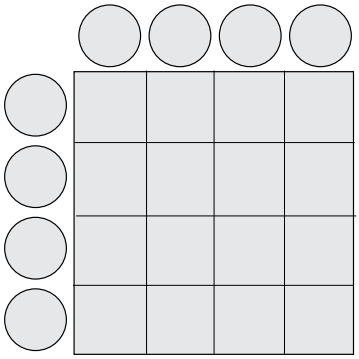
Genotype ratio: _____

_____

Phenotype ratio: _____

_____

_____





(b) A tabby, short haired male (**TtbLl**) is crossed with a blotched tabby, short haired (**tbtbLl**) female. Determine ratios of the offspring:

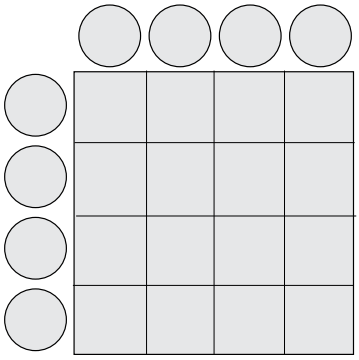
Genotype ratio: _____

_____

Phenotype ratio: _____

_____

_____



2. A plant with orange-striped flowers was cultivated from seeds. The plant was self-pollinated and the F_1 progeny appeared in the following ratios: 89 orange with stripes, 29 yellow with stripes, 32 orange without stripes, 9 yellow without stripes.

(a) Describe the dominance relationships of the alleles responsible for the phenotypes observed: _____

_____

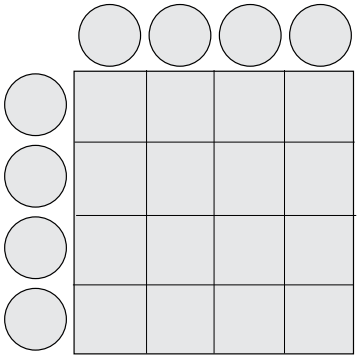
_____

(b) Determine the genotype of the original plant with orange striped flowers: _____

_____

_____

3. In rabbits, spotted coat **S** is dominant to solid colour **s**, while for coat colour, black **B** is dominant to brown **b**. A brown spotted rabbit is mated with a solid black one and all the offspring are black spotted (the genes are not linked).



(a) State the genotypes:

Parent 1: _____

Parent 2: _____

Offspring: _____

(b) Use the Punnett square to show the outcome of a cross between the F_1 (the F_2):

(c) Using ratios, state the phenotypes of the F_2 generation: _____

_____

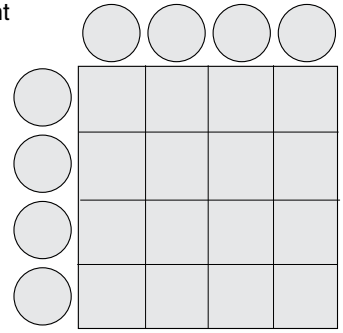
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LINK 189 TEST

4. In guinea pigs, rough coat **R** is dominant over smooth coat **r** and black coat **B** is dominant over white **b**. The genes are not linked.
A homozygous rough black animal was crossed with a homozygous smooth white:



(a) State the genotype of the F_1: _____

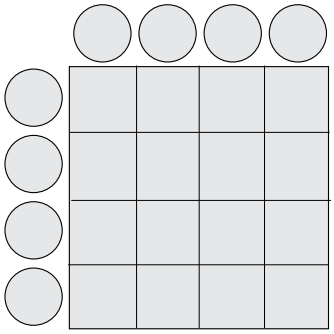
(b) State the phenotype of the F_1: _____

(c) Use the Punnett square (top right) to show the outcome of a cross between the F_1 (the F_2):

(d) Using ratios, state the phenotypes of the F_2 generation: _____

_____

_____



(e) Use the Punnett square (right) to show the outcome of a **back cross** of the F_1 to the rough, black parent:

(f) Using ratios, state the phenotype of the F_2 generation: _____

_____

(g) A rough black guinea pig was crossed with a rough white one produced the following offspring: 28 rough black, 31 rough white, 11 smooth black, and 10 smooth white. Determine the genotypes of the parents:

_____

5. The Himalayan colour-pointed, long-haired cat is a breed developed by crossing a pedigree (true-breeding), uniform-coloured, long-haired Persian with a pedigree colour-pointed (darker face, ears, paws, and tail) short-haired Siamese.


Persian


Siamese


Himalayan

The genes controlling hair colouring and length are on separate chromosomes: uniform colour **U**, colour pointed **u**, short hair **S**, long hair **s**.

(a) Using the symbols above, indicate the genotype of each breed below its photograph (above, right). _____ _____ _____

(b) State the genotype of the F_1 (Siamese X Persian): _____

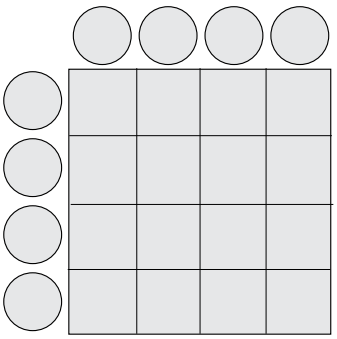
(c) State the phenotype of the F_1: _____

(d) Use the Punnett square to show the outcome of a cross between the F_1 (the F_2):

(e) State the ratio of the F_2 that would be Himalayan: _____

(f) State whether the Himalayan would be true breeding: _____

(g) State the ratio of the F_2 that would be colour-point, short-haired cats: _____



6. A *Drosophila* male with genotype **Cucu Ebeb** (straight wing, grey body) is crossed with a female with genotype **cucu ebeb** (curled wing, ebony body). The phenotypes of the F_1 were recorded and the percentage of each type calculated. The percentages were: Straight wings, grey body 45%, curled wings, ebony body 43%, straight wings, ebony body 6%, and curled wings grey body 6%.


Straight wing Cucu Grey body, Ebeb


Curled wing cucu Ebony body, ebeb

(a) Is there evidence of crossing over in the offspring? _____

(b) Explain your answer: _____

_____

_____

(c) Determine the genotypes of the offspring: _____

_____

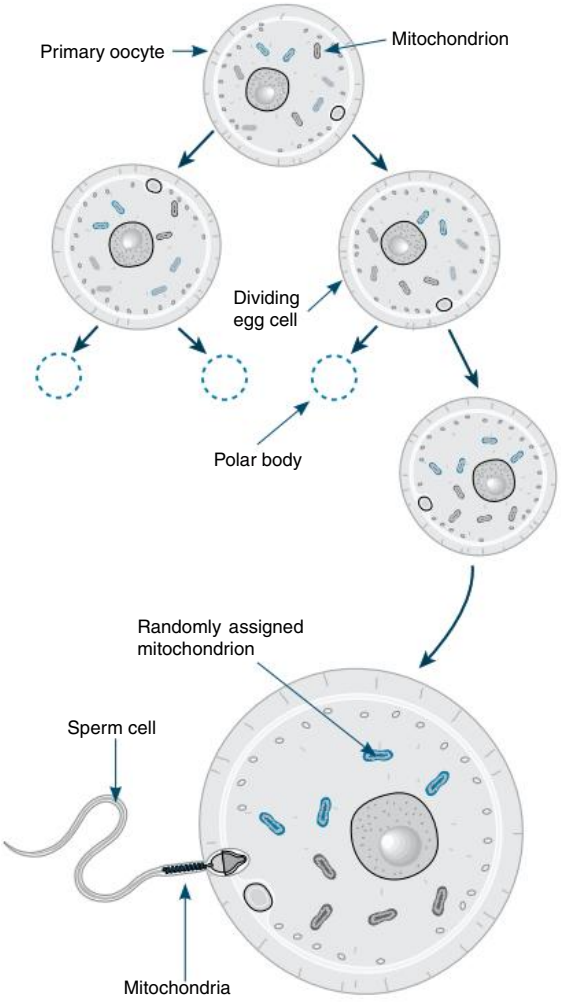
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194 Non-nuclear Inheritance

Key Idea: Not all genes are encoded within nuclear DNA. Both mitochondria and chloroplasts contain their own plasmid-like copies of DNA, (denoted mtDNA and cpDNA respectively). Mitochondrial and chloroplast DNA replicates independently of the main genome and contains genes essential for that organelle's function, e.g. mitochondrial DNA contains genes for producing some of the proteins associated with the electron transport chain. Because mitochondria and chloroplasts are distributed randomly in gametes, traits determined by their genes do not follow simple Mendelian rules.

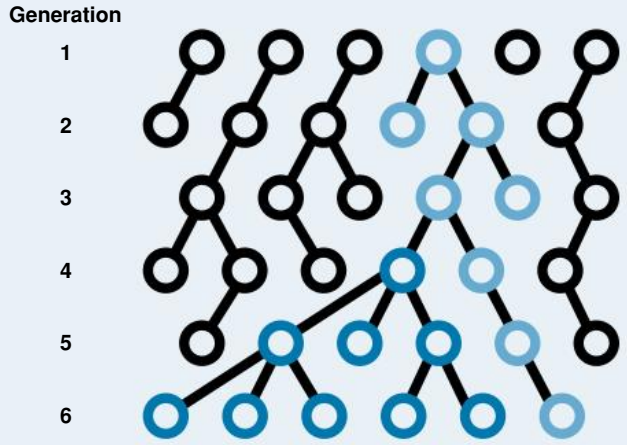
Mitochondrial and Chloroplast DNA

In most multicellular organisms, the mtDNA and cpDNA are inherited from the maternal line. Mitochondria in the cytoplasm are randomly divided between dividing primary oocytes so that the final egg cell has a random selection of mitochondria from the original cell. Mitochondria are numerous in sperm cells, providing the energy to power the tail. However, they do not enter the egg cell, or are destroyed by enzymes within it.



Using mtDNA and cpDNA

Because mitochondria and chloroplasts are passed through the maternal line they can be used to trace maternal lineage. Mutations occur in mtDNA at a rate of fewer than one per 100 people. This means that the mtDNA of any one person is probably the same as their direct maternal ancestor back many generations. The same applies for plant cpDNA and mtDNA. In humans, this concept has been used to trace our mitochondrial ancestor, a single female from Africa. This does not mean this person is the ancestor of all humans, rather that no other lineages produced daughters through which the mtDNA was passed to our generation.



Experimental Evidence for Maternal Inheritance of cpDNA



In 1909, Carl Correns, a German geneticist, noticed that the branches of the four o'clock (*Mirabilis jalapa*) developed leaves that could be green, white, or variegated (a combination of green and white). After carrying out a series of crosses, Correns noticed that seeds from branches with solid green leaves grew into plants with solid green leaves, even if the pollen was from a branch with white or variegated leaves. Seeds from branches with variegated leaves produced plants with all three leaf types. Again, where the pollen came from did not matter. In addition, the appearance of these three types was never in any predictable Mendelian ratio. Correns concluded that the trait was being passed via the maternal line. We now know a defect in the cpDNA of the four o'clock causes the white colour and these, like mitochondria, are passed randomly to the offspring.

1. (a) Why are mtDNA and cpDNA only passed along the maternal line? _____

_____

_____

(b) Why do the inheritance patterns of mtDNA and cpDNA not follow predictable Mendelian rules? _____

_____

2. (a) In which generation of the mtDNA diagram (blue box above) did a mutation in the mtDNA occur? _____

(b) How can this be used to help trace the lineage? _____

_____

_____

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195 DNA Methylation

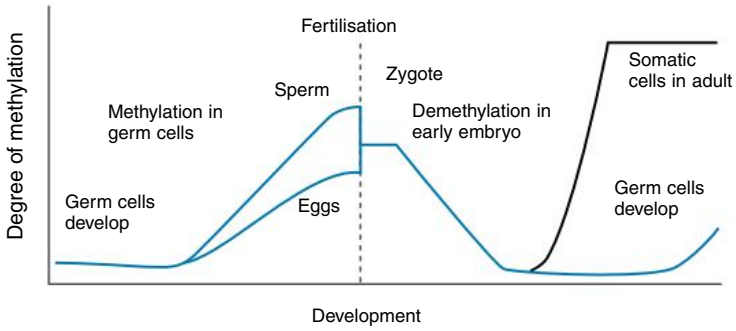
Key Idea: Methylation of the DNA can alter gene expression and therefore phenotype.

Methylation of DNA is an important way of controlling gene expression. Methylated DNA is usually silenced, meaning genes are not transcribed to mRNA. Methylation of cytosine turns off gene expression by changing the state of the chromatin so that transcribing proteins are not able to bind to the DNA. Methylation is also important in X-inactivation.

Methylation and Gene Expression

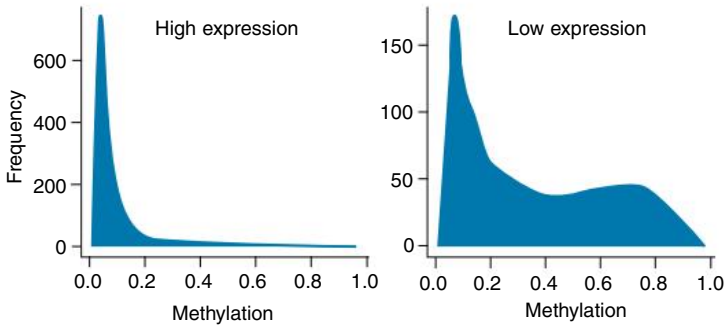
Gene expression changes as an organism develops. During the development of the embryo there are many genes that are switched on and off as development of tissues and organs proceeds. Some of this control is achieved by DNA methylation. Enzymes can add or remove methyl groups from cytosine bases and so activate or silence genes. Often methylation is affected by changes in the environment and this provides the developing organism with a rapid response mechanism.

Methylation in Mammalian Development



Methylation vs Gene Expression

A comparison of methylation and gene expression finds that highly expressed genes have very little methylation while genes that are not expressed have very high levels of methylation.



Methylation and Imprinted Genes

Genomic imprinting is a phenomenon in which the pattern of gene expression is different depending on whether the gene comes from the mother or the father. Imprinted genes are silenced by methylation and histone modification. A gene inherited from the father may be silenced while the gene inherited from the mother may be active or vice versa. Evidence of this is seen in two human genetic disorders, Angelman syndrome and Prader-Willi syndrome. Both are caused by the same mutation; a specific deletion on chromosome 15. Which syndrome is expressed depends on whether the mutation occurs on the maternal or paternal chromosome.


A liger

The effect of genomic imprinting can be seen in other mammals. Ligers (a cross between a male lion and a female tiger) although not occurring in the wild, are the biggest of the big cats. However a tigon (a cross between a female lion and a male tiger) is no bigger than a normal lion. It is thought this difference in phenotype is due to the male lion carrying imprinted genes that result in larger offspring which are normally counteracted by genes from the female. Similarly the differences between a mule (male donkey + female horse) and a hinny (female donkey + male horse) may be to do with genomic imprinting.

1. (a) What is DNA methylation? _____

   _____

   (b) How does DNA methylation affect gene expression?: _____

   _____

   _____

2. Prader-Willi syndrome is caused when a mutated gene on chromosome 15 is inherited from the father. How does this tell us that the mother must therefore have donated the imprinted gene?

   _____

   _____

   _____

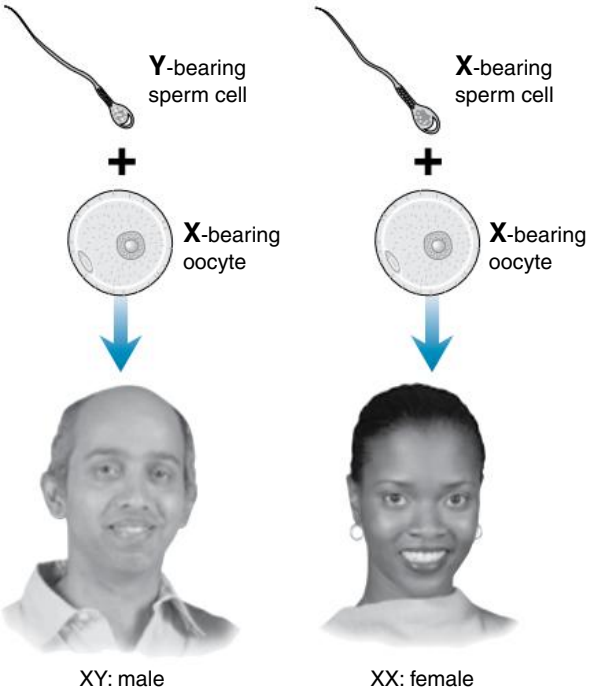
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196 Sex Determination

Key Idea: The sex of an individual is usually determined by sex chromosomes, but can be influenced by the environment.

The determination of sex in an organism is controlled in most cases by the sex chromosomes provided by each parent. These have evolved to regulate the ratios of males and females produced and preserve the genetic differences between the sexes. In humans, males are the **heterogametic sex** because each somatic cell has one X and one Y chromosome. Sex determination is based on the presence or absence of the Y chromosome. Without a Y, an individual will be female. In mammals, the male is always the heterogametic sex, but this is not necessarily the case in other taxa. In birds and butterflies, the female is the heterogametic sex, and in some insects the male is simply X whereas the female is XX.

Sex Determination in Humans

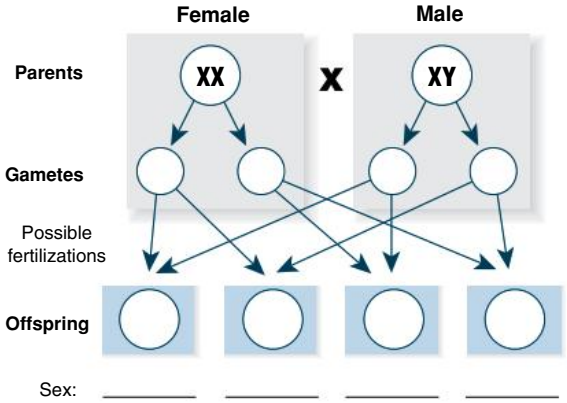


XY Sex Determination

Female: **XX** Male: **XY**

Examples: Humans (and all mammals), fruit flies (*Drosophila*), some dioecious plants (having separate male and female plants) such as kiwifruit.

In humans the female is the **homogametic sex** and has two similar sex chromosomes (XX), whereas the male is the heterogametic sex with two unlike chromosomes (XY). The primary sexual characteristics are initiated by special genes on the X chromosomes. Females must have a double dose (2X chromosomes). Maleness is determined by the presence of the Y chromosome.



Sex Determination in *Drosophila*



In *Drosophila*, maleness is determined by the ratio of X chromosomes to autosomes, not the presence of the Y chromosome. Two X chromosomes in a diploid cell produce a female fly, but one X chromosome in a diploid cell produces a male fly. The Y chromosome is not involved in determining sex, but does contains genes involved in the production of sperm in adult males. Thus XO in *Drosophila* is a sterile male, while in mammals it is a sterile female.

Temperature Dependent Sex Determination



In some vertebrate species, mostly reptiles, sex is determined by the temperature at which the eggs are incubated. In turtles, males are produced at lower incubation temperatures than females (22°C-27°C as opposed to 30°C). The hormone testosterone may be converted to estradiol at higher temperatures to produce females, but how temperature triggers gene expression and the pathway for genetic sex determination is poorly understood.

1. (a) Complete the diagram above, to show the resulting gametes, genotype and sex of the offspring:

   (b) Determine the probability of a conception producing a male child: _____

   (c) Determine the probability of a second conception producing a female child: _____

2. Explain what determines the sex of the offspring at the moment of conception in humans: _____

   _____

3. Explain why many genes on the X chromosome in males will be expressed regardless of their dominance status:

   _____

   _____

4. Estimate the ratio of males to females produced when a clutch of turtle eggs is incubated at 28.5°C: _____

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197 Sex Linked Genes

Key Idea: Many genes on the X chromosome do not have a match on the Y chromosome. In males, a recessive allele cannot therefore be masked by a dominant allele.

Sex linkage occurs when a gene is located on a sex chromosome (usually the X). The result of this is that the character encoded by the gene is usually seen only in one sex (the heterogametic sex). In humans, recessive sex linked genes cause a number of heritable disorders in males, e.g. haemophilia. Women who have a recessive allele are said to be **carriers**. One of the gene loci controlling coat colour in cats is sex-linked. The two alleles, red and non-red (or black), are found only on the X-chromosome.

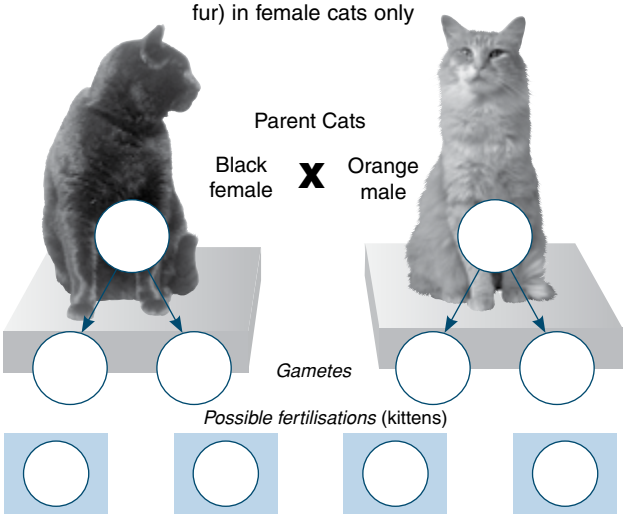


Allele types			Genotypes		Phenotypes
X_o	=	Non-red (=black)	X_oX_o, X_oY	=	Black coated female, male
X_O	=	Red	X_OX_O, X_OY	=	Orange coated female, male
			X_OX_o	=	Tortoiseshell (intermingled black and orange in fur) in female cats only

1. An owner of a cat is thinking of mating her black female cat with an orange male cat. Before she does this, she would like to know what possible coat colours could result from such a cross. Use the symbols above to fill in the diagram on the right. Summarise the possible genotypes and phenotypes of the kittens in the tables below.

	Genotypes	Phenotypes
Male kittens		

Female kittens		

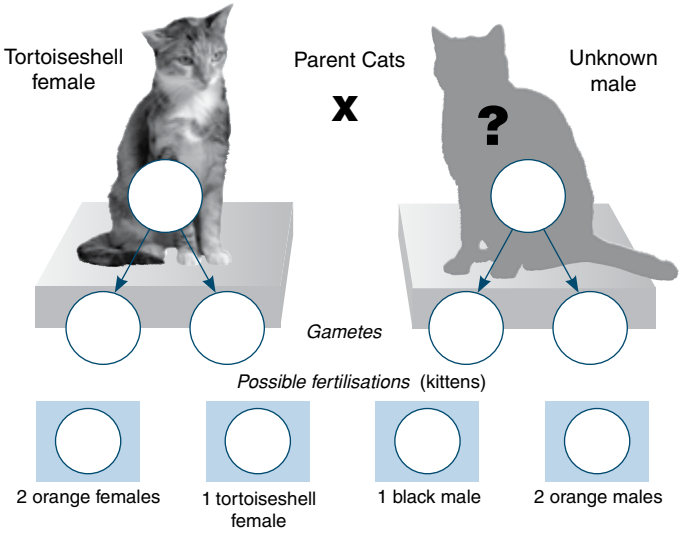


2. A female tortoiseshell cat mated with an unknown male cat in the neighbourhood and has given birth to a litter of six kittens. The owner of this female cat wants to know what the appearance and the genotype of the father was of these kittens. Use the symbols above to fill in the diagram on the right. Also show the possible fertilisations by placing appropriate arrows.

   Describe the father cat's:

   (a) Genotype: _____

   (b) Phenotype: _____



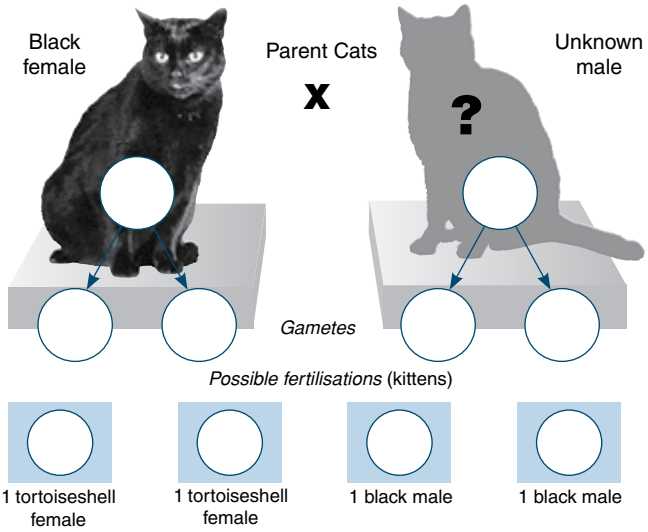
3. The owner of another cat, a black female, also wants to know which cat fathered her two tortoiseshell female and two black male kittens. Use the symbols above to fill in the diagram on the right. Show the possible fertilisations by placing appropriate arrows.

   Describe the father cat's:

   (a) Genotype: _____

   (b) Phenotype: _____

   (c) Was it the same male cat that fathered both this litter and the one above?
   YES / **NO** (*delete one*)



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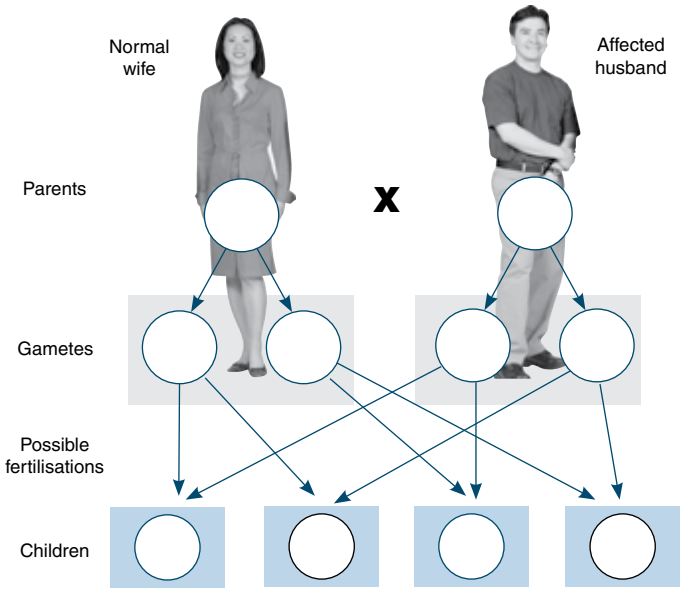


Dominant allele in humans

A rare form of rickets in humans is determined by a **dominant** allele of a gene on the **X chromosome** (it is not found on the Y chromosome). This condition is not successfully treated with vitamin D therapy. The allele types, genotypes, and phenotypes are as follows:

Allele types		Genotypes		Phenotypes
X_R	= affected by rickets	X_RX_R, X_RX	=	Affected female
X	= normal	X_RY	=	Affected male
		XX, XY	=	Normal female, male

As a genetic counselor you are presented with a married couple where one of them has a family history of this disease. The husband is affected by this disease and the wife is normal. The couple, who are thinking of starting a family, would like to know what their chances are of having a child born with this condition. They would also like to know what the probabilities are of having an affected boy or affected girl. Use the symbols above to complete the diagram right and determine the probabilities stated below (expressed as a proportion or percentage).



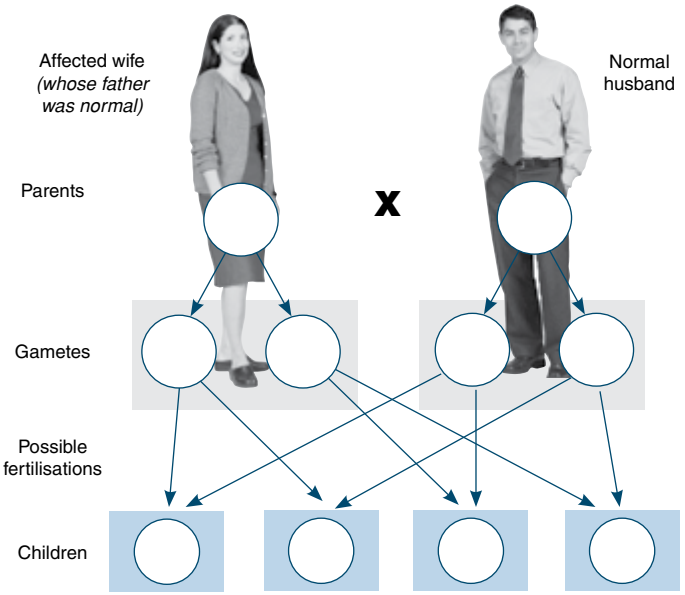
4. Determine the probability of having:

   (a) Affected children: _____

   (b) An affected girl: _____

   (c) An affected boy: _____

Another couple with a family history of the same disease also come in to see you to obtain genetic counselling. In this case the husband is normal and the wife is affected. The wife's father was not affected by this disease. Determine what their chances are of having a child born with this condition. They would also like to know what the probabilities are of having an affected boy or affected girl. Use the symbols above to complete the diagram right and determine the probabilities stated below (expressed as a proportion or percentage).



5. Determine the probability of having:

   (a) Affected children: _____

   (b) An affected girl: _____

   (c) An affected boy: _____

6. Describing examples other than those above, discuss the role of **sex linkage** in the inheritance of genetic disorders:

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198 Inheritance Patterns

Key Idea: Sex-linked traits and autosomal traits have different inheritance patterns.

Complete the following monohybrid crosses for different types of inheritance patterns in humans: autosomal recessive, autosomal dominant, sex linked recessive, and sex linked dominant inheritance.

1. **Inheritance of autosomal recessive traits**
   Example: *Albinism*

   Albinism (lack of pigment in hair, eyes and skin) is inherited as an autosomal recessive allele (not sex-linked).

   Using the codes: **PP** (normal) **Pp** (carrier)
   pp (albino)

   (a) Enter the parent phenotypes and complete the Punnett square for a cross between two carrier genotypes.

   (b) Give the ratios for the phenotypes from this cross.

   Phenotype ratios: _____

   _____

2. **Inheritance of autosomal dominant traits**
   Example: *Woolly hair*

   Woolly hair is inherited as an autosomal dominant allele. Each affected individual will have at least one affected parent.

   Using the codes: **WW** (woolly hair)
   Ww (woolly hair, heterozygous)
   ww (normal hair)

   (a) Enter the parent phenotypes and complete the Punnett square for a cross between two heterozygous individuals.

   (b) Give the ratios for the phenotypes from this cross.

   Phenotype ratios: _____

   _____

3. **Inheritance of sex linked recessive traits**
   Example: *Hemophilia*

   Inheritance of hemophilia is sex linked. Males with the recessive (hemophilia) allele, are affected. Females can be carriers.

   Using the codes: **XX** (normal female)
   XX_h (carrier female)
   X_hX_h (hemophiliac female)
   XY (normal male)
   X_hY (hemophiliac male)

   (a) Enter the parent phenotypes and complete the Punnett square for a cross between a normal male and a carrier female.

   (b) Give the ratios for the phenotypes from this cross.

   Phenotype ratios: _____

   _____

4. **Inheritance of sex linked dominant traits**
   Example: *Sex linked form of rickets*

   A rare form of rickets is inherited on the X chromosome.

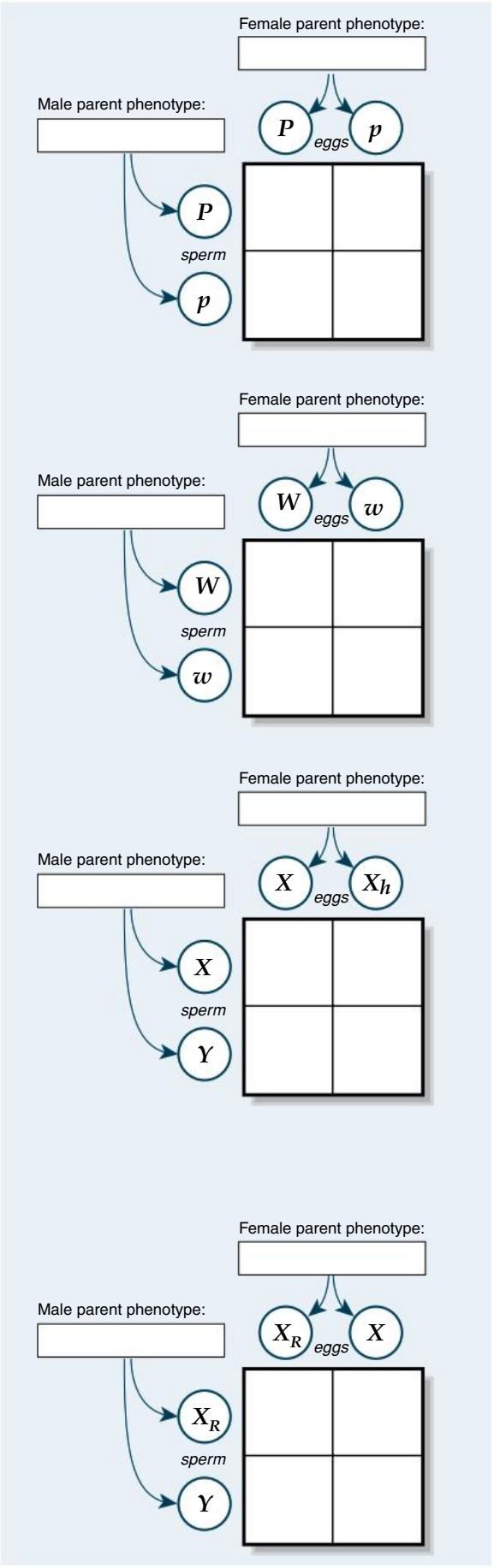
   Using the codes: **XX** (normal female); **XY** (normal male)
   X_RX (affected heterozygote female)
   X_RX_R (affected female)
   X_RY (affected male)

   (a) Enter the parent phenotypes and complete the Punnett square for a cross between an affected male and heterozygous female.

   (b) Give the ratios for the phenotypes from this cross.

   Phenotype ratios: _____

   _____

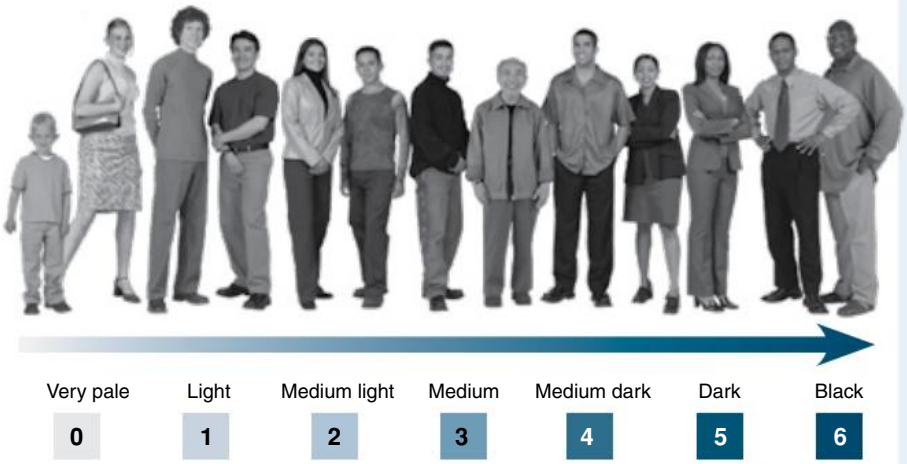


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199 Polygenes

Key Idea: Many phenotypes are affected by multiple genes.

Some phenotypes (e.g. kernel colour in maize and skin colour in humans) are determined by more than one gene and show **continuous variation** in a population. The production of the skin pigment melanin in humans is controlled by at least three genes. The amount of melanin produced is directly proportional to the number of dominant alleles for either gene (from 0 to 6).




A light-skinned person


A dark-skinned person

There are seven shades skin colour ranging from very dark to very pale, with most individual being somewhat intermediate in skin colour. No dominant allele results in a lack of dark pigment (aabbcc). Full pigmentation (black) requires six dominant alleles (AABBCC).

1. Complete the Punnett square for the F_2 generation (below) by entering the genotypes and the number of dark alleles resulting from a cross between two individuals of intermediate skin colour. Colour-code the offspring appropriately for easy reference.

   (a) How many of the 64 possible offspring of this cross will have darker skin than their parents:

   _____

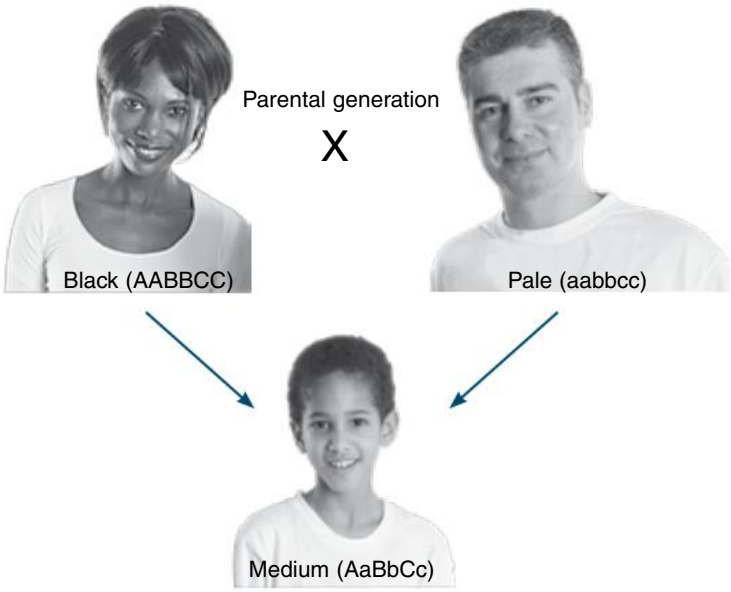
   (b) How many genotypes are possible for this type of gene interaction:

   _____

2. Explain why we see many more than seven shades of skin colour in reality:

   _____

   _____



F2 generation (AaBbCc X AaBbCc)

GAMETES	ABC	ABc	AbC	Abc	aBC	aBc	abC	abc
ABC								
ABc								
AbC								
Abc								
aBC								
aBc								
abC								
abc								

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3. Discuss the differences between **continuous** and **discontinuous** variation, giving examples to illustrate your answer:

4 From a sample of no less than 30 adults, collect data (by request or measurement) for one continuous variable (e.g. height, weight, shoe size, or hand span). Record and tabulate your results in the space below, and then plot a frequency histogram of the data on the grid below:



Raw data

Tally Chart (frequency table)

Variable: .....

Frequency

(a) Calculate the following for your data and attach your working. See the Weblink: *Descriptive Statistics* if you need help:

Mean: _____ **Mode**: _____ **Median**: _____

Standard deviation: _____

(b) Describe the pattern of distribution shown by the graph, giving a reason for your answer: _____

(c) What is the genetic basis of this distribution? _____

(d) What is the importance of a large sample size when gathering data relating to a continuous variable?

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200 Pedigree Analysis

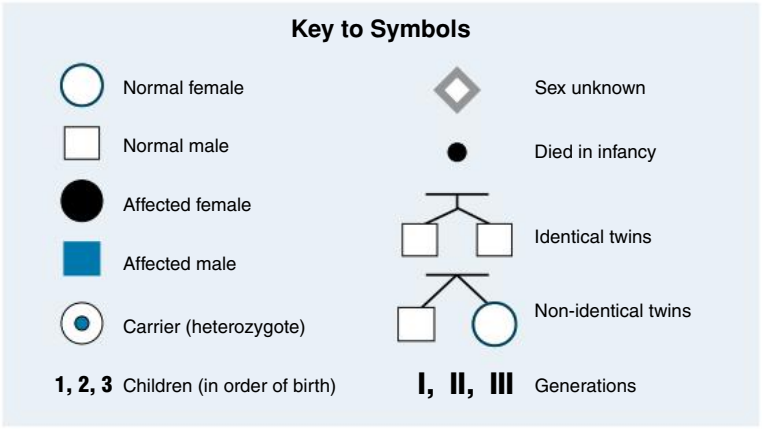
Key Idea: Pedigree charts are a way to illustrate inheritance patterns over a number of generations graphically. They are used to study the inheritance of genetic disorders and make it possible to follow the genetic history of an individual.

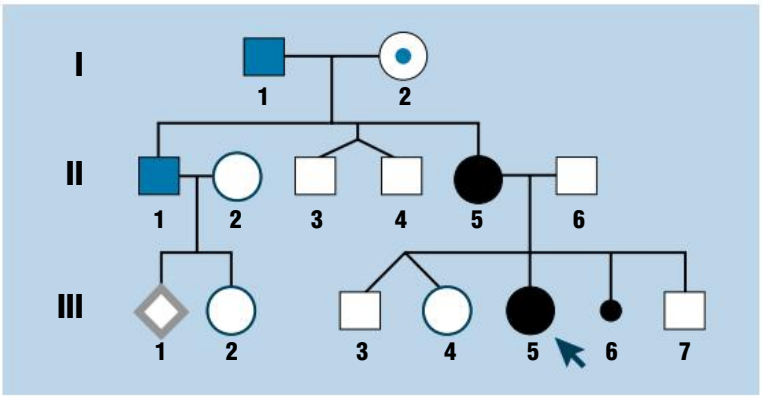
Sample Pedigree Chart

Pedigree charts use various symbols to indicate an individuals particular traits. The key (right) should be consulted to make sense of the various symbols. Particular individuals are identified by their generation number and their order number in that generation. For example, **II-6** is the sixth person in the second row. The arrow indicates the **propositus**; the person through whom the pedigree was discovered (i.e. who reported the condition).

If the chart on the right were illustrating a human family tree, it would represent three generations: grandparents (I-1 and I-2) with three sons and one daughter. Two of the sons (II-3 and II-4) are identical twins, but did not marry or have any children. The other son (II-1) married and had a daughter and another child (sex unknown). The daughter (II-5) married and had two sons and two daughters (plus a child that died in infancy).

For the particular trait being studied, the grandfather was expressing the phenotype (showing the trait) and the grandmother was a carrier. One of their sons and one of their daughters also show the trait, together with one of their granddaughters.





1. **Pedigree chart of your family**
   Using the symbols in the key above and the example illustrated as a guide, construct a pedigree chart of your own family (or one that you know of) starting with the parents of your mother and/or father on the first line. Your parents will appear on the second line (II) and you will appear on the third line (III). There may be a fourth generation line (IV) if one of your brothers or sisters has had a child. Use a ruler to draw up the chart carefully.

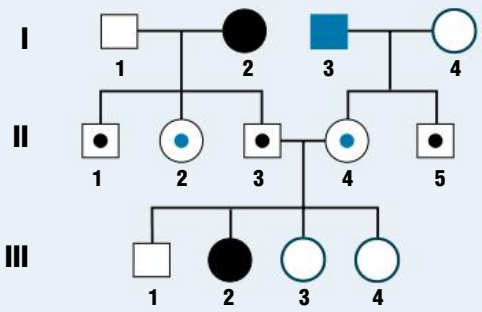
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2. **Autosomal recessive traits**
Albinos lack pigment in the hair, skin and eyes. This trait is inherited as an autosomal recessive allele (i.e. it is not carried on the sex chromosome).

(a) Write the genotype for each of the individuals on the chart using the following letter codes: **PP** normal skin colour; **P-** normal, but unknown if homozygous; **Pp** carrier; **pp** albino.

(b) Why must the parents (II-3) and (II-4) be **carriers** of a **recessive** allele:

Albinism in humans

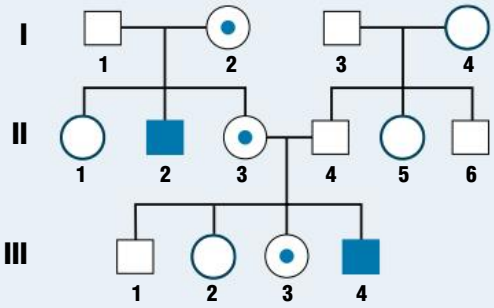


3. **Sex linked recessive traits**
Haemophilia is a disease where blood clotting is affected. A person can die from a simple bruise (which is internal bleeding). The clotting factor gene is carried on the X chromosome.

(a) Write the genotype for each of the individuals on the chart using the codes: **XY** normal male; **X_hY** affected male; **XX** normal female; **X_hX** female carrier; **X_hX_h** affected female:

(b) Why can males never be carriers?

Haemophilia in humans

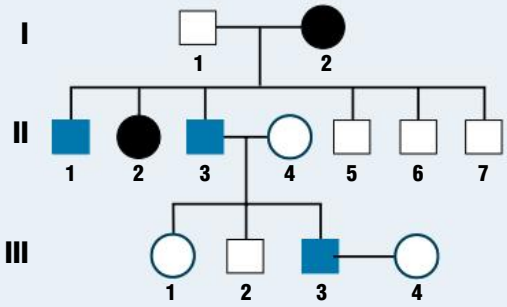


4. **Autosomal dominant traits**
An unusual trait found in some humans is woolly hair (not to be confused with curly hair). Each affected individual will have at least one affected parent.

(a) Write the genotype for each of the individuals on the chart using the following letter codes:
WW woolly hair; **Ww** woolly hair (heterozygous); **W-** woolly hair, but unknown if homozygous; **ww** normal hair

(b) Describe a feature of this inheritance pattern that suggests the trait is the result of a **dominant** allele:

Woolly hair in humans

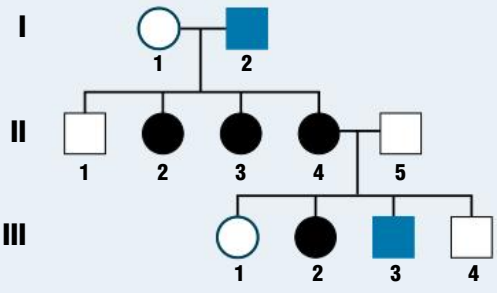


5. **Sex linked dominant traits**
A rare form of rickets is inherited on the X chromosome. All daughters of affected males will be affected. More females than males will show the trait.

(a) Write the genotype for each of the individuals on the chart using the following letter codes:
XY normal male; **X_RY** affected male; **XX** normal female; **X_{R-}** female (unknown if homozygous); **X_RX_R** affected female.

(b) Why will more females than males be affected?

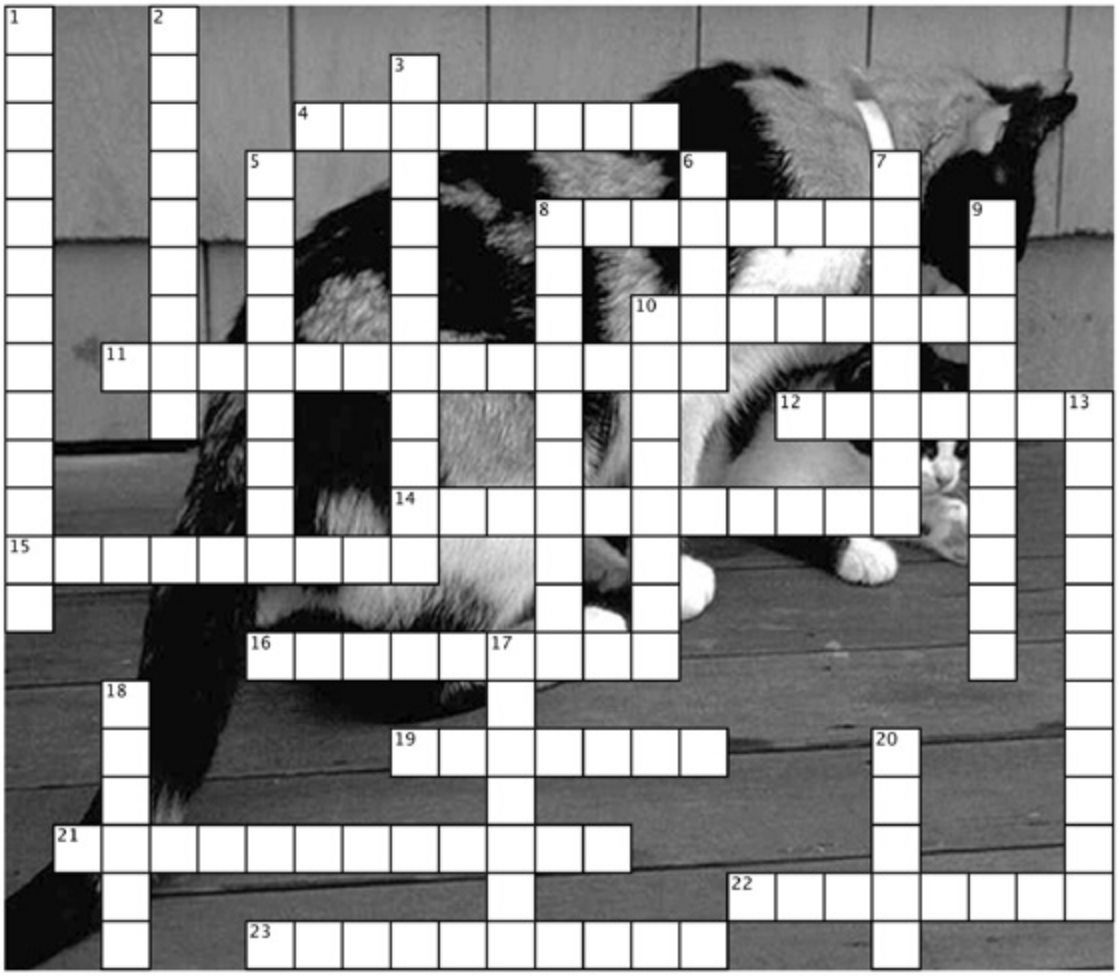
A rare form of rickets in humans



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201 KEY TERMS: Crossword

Complete the crossword below to test your understanding of key terms in this chapter and their meanings



Clues Across

4. The specific allele combination of an organism.
8. A chromosome that is not a sex chromosome.
10. An alteration in the base sequence of DNA.
11. Variation that falls into discrete groups.
12. An individual with a single recessive allele for a condition that requires a dominant allele for expression.
14. An inheritance patten where both alleles in the heterozygous condition contribute to the phenotype.
15. Allele that expresses its characteristics only when in the homozygous condition.
16. The physical appearance of the genotype.
19. A reduction division responsible for the production of haploid gametes or spores.
21. Possessing two different alleles at a locus.
22. The allele that expresses its characteristics when in both homozygous and heterozygous conditions.
23. Possessing two alleles at a locus that are the same.

Clues Down

1. A diagram that is used to predict an outcome of a particular genetic cross (2 words: 7, 6)
2. Characteristics that are a product of multiple genes being expressed are called _____ .
3. The acquisition of characteristics by the transfer of genetic information from one generation to the next.
5. A diagnostic cross involving breeding with a recessive with known genotype. (2 words: 4, 5)
6. The position of a gene on a chromosome.
7. The family history or inheritance of an organism is often referred to as its _____
8. An abnormal condition in which the chromosome number is not an exact multiple of the normal haploid set for the species.
9. Variation that exhibits a wide range of graduated phenotypes is called this.
10. Genes with more than two types of alleles are called _____ alleles.
13. Offspring of parents with linked genes that do not display the parental phenotype are called _____ offspring.
17. The form of polysomy in which one chromosomal pair is represented by three chromosomes instead of two.
18. One of two or more forms of a particular gene.
20. A particular phenotypic character. Refers to the physical appearance as opposed to the mode of appearance, e.g. blue eyes rather than eye colour.

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202 Heredity Synoptic Question

1. What is the purpose of DNA replication? _____

2. Explain how the parent DNA is copied by semi-conservative replication (you may use extra paper if required).
   Your answer should include:
   - the units that make up DNA
   - the process of replication
   - a labelled diagram to support your answer

3. In guinea pigs, black hair (B) is dominant over white hair (b) and short hair (L) is dominant over long hair (l). In order to produce consistent and reliable coat colour in guinea pigs, breeders use true breeding parents. A breeder has a male guinea she knows to be BBLL and a female she knows to be bbll. She wants to use them to begin a breeding programme to produce a pure breeding white, short haired guinea pig.

   Explain how the desired pure breeding animal can be produced and how its ability to breed true can be proved. Support your answer with Punnett squares and the correct use of the allele letters given (you may use extra paper if required):

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QLD As required
VCE Unit 4 (2)
WA ATAR Unit 3
SA Theme E

Population Genetics

Key terms

adaptation
allele
allele frequency
Darwin
deme
differential survival
directional selection
disruptive selection
evolution
fitness
fixation (of alleles)
founder effect
gene flow
gene pool
genetic bottleneck
genetic drift
genetic equilibrium
genotype
heterozygous advantage
mate choice
microevolution
migration
mutation
natural selection
new synthesis
phenotype
polymorphism
population
species
stabilising selection
Wallace

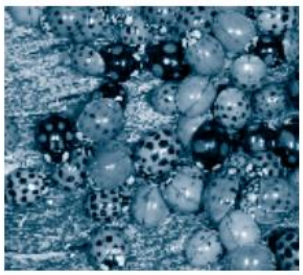
The modern synthesis of evolution

Learning aims and skills

Activity number

- ☐ 1 Explain what is meant by biological evolution. Describe the new synthesis of evolution that takes into account our current understanding of molecular genetics and inheritance. — 203 204
- ☐ 2 Outline the fundamental ideas in Darwin's "Theory of evolution by natural selection" and their significance. — 204
- ☐ 3 Explain the term fitness. Explain how evolution, through adaptation, equips species for survival. Describe adaptations of organisms to their environment. — 209





Species and gene pools

Learning aims and skills

Activity number

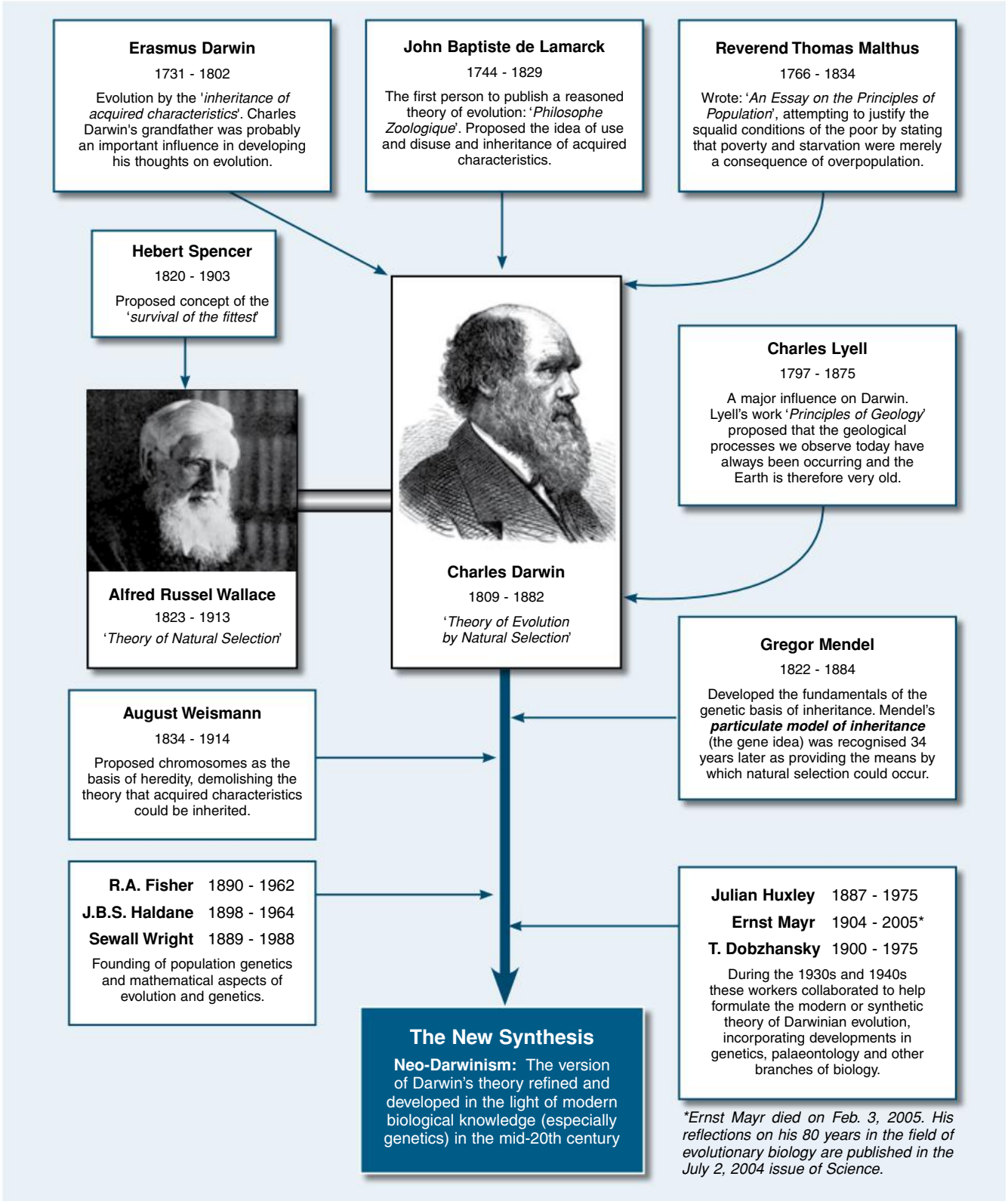
- ☐ 4 Understand the concept of the gene pool and explain the term deme. — 206
- ☐ 5 Explain how allele frequencies are expressed for populations. Explain the effect of mutation, gene flow (migration), natural selection, and genetic drift on the allele frequencies of populations. — 206
- ☐ 6 State the conditions required for genetic equilibrium in a population and explain the consequences of these conditions rarely, if ever, being satisfied. — 206
- ☐ 7 Explain the role of natural selection in sorting the variability within a gene pool and establishing adaptive genotypes. — 206-209
- ☐ 8 Distinguish between stabilising, disruptive, and directional selection. — 208
- ☐ 9 Describe and explain examples of stabilising, disruptive, and directional selection. Selection for human skin colour represents an interesting case of latitude dependent stabilising selection (the adaptive phenotype varies depending on the latitude). — 210-214
- ☐ 10 Describe and evaluate examples of genetic change in real populations over time. Suitable examples include selection for beak size in Darwin's finches (directional on the island of Daphne major and directional on the island Santa Cruz), industrial melanism in peppered moths in the UK, heterozygous advantage and the sickle cell gene, antibiotic resistance in bacteria, or pesticide resistance in insects. Appreciate that evolution in viruses (e.g. *Influenzavirus*) is a function of their high mutation rates and the selective environment represents the host's immune response. — 211-213 215-218
- ☐ 11 Explain the genetic and evolutionary consequences of the founder effect. — 219
- ☐ 12 Explain the genetic and evolutionary consequences of the bottleneck effect. — 220
- ☐ 13 Recognise genetic drift as an important process in evolution. Describe its consequences and the conditions under which it is important. — 221



203 The Modern Theory of Evolution

Key Idea: Charles Darwin is credited with developing the theory of evolution by natural selection, but the contributions of others were important to the formulation of his ideas.

The development of the modern theory of evolution has a history going back at least two centuries. The diagram below summarises how some of the major contributors helped to form the currently accepted model, or **new synthesis**. Our understanding of evolutionary processes expanded through the 1980s and 1990s as comparative molecular sequence data were gathered and understanding of the molecular basis of development improved. More recently, evolutionary developmental biology (**evo-devo**) is helping us to understand how changes in gene regulation during development can result in the development of novel characteristics.



1. From the diagram above, choose one of the contributors to the development of evolutionary theory (excluding Charles Darwin himself), and write a few paragraphs discussing their role in contributing to Darwin's ideas. You may need to consult an external reference source for help.

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204 Mechanism of Natural Selection

Key Idea: Natural selection is the evolutionary mechanism by which organisms that are better adapted to their environment survive to produce a greater number of offspring.

Evolution is the change in inherited characteristics in a population over generations. Evolution is the consequence of interaction between four factors: (1) The potential for populations to increase in numbers, (2) Genetic variation as a result of mutation and sexual reproduction, (3) competition for resources, and (4) proliferation of individuals with better survival and reproduction.

Natural selection is the term for the mechanism by which better adapted organisms survive to produce a greater number of viable offspring. This has the effect of increasing their proportion in the population so that they become more common. This is the basis of Darwin's theory of evolution by natural selection.

We can demonstrate the basic principles of evolution using the analogy of a 'population' of M&M's candy.



In a bag of M&M's, there are many colours, which represents the variation in a population. As you and a friend eat through the bag of candy, you both leave the blue ones, which you both dislike, and return them to bag.



The blue candy becomes more common...



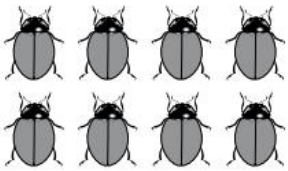
Eventually, you are left with a bag of blue M&M's. Your selective preference for the other colours changed the make-up of the M&M's population. This is the basic principle of selection that drives evolution in natural populations.

Darwin's Theory of Evolution by Natural Selection

Darwin's theory of evolution by natural selection is outlined below. It is widely accepted by the scientific community today and is one of founding principles of modern science.

Overproduction

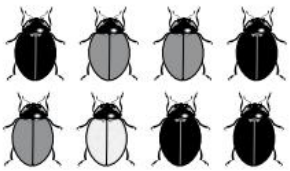
Populations produce too many young: many must die



Populations generally produce more offspring than are needed to replace the parents. Natural populations normally maintain constant numbers. A certain number will die without reproducing.

Variation

Individuals show variation: some variations more favourable than others



Individuals in a population have different phenotypes and therefore, genotypes. Some traits are better suited to the environment, and individuals with these have better survival and reproductive success.

Natural Selection

Natural selection favours the individuals best suited to the environment at the time



Individuals in the population compete for limited resources. Those with favourable variations will be more likely to survive. Relatively more of those without favourable variations will die.

Inherited

Variations are inherited:
the best suited variants leave more offspring

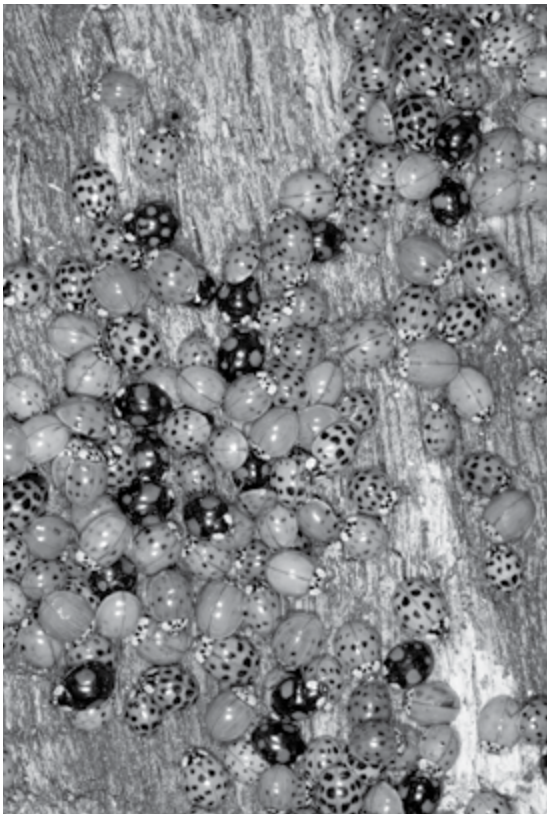


The variations (both favourable and unfavourable) are passed on to offspring. Each generation will contain proportionally more descendants of individuals with favourable characters.

1. Identify the four factors that interact to bring about evolution in populations: _____

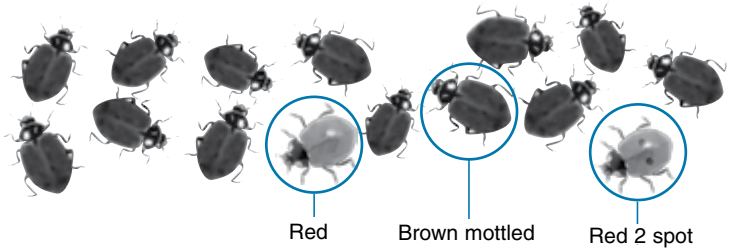
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Variation, Selection, and Population Change

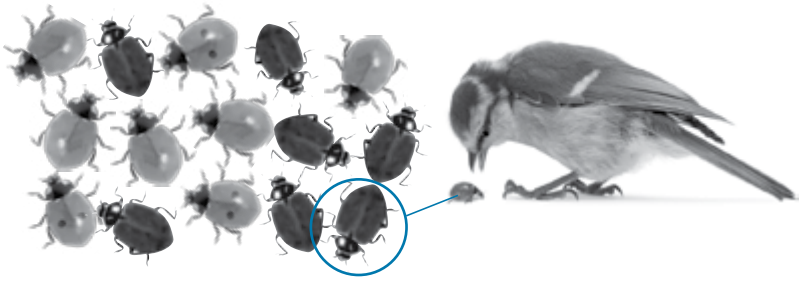


Natural populations, like the ladybug population above, show genetic variation. This is a result of **mutation** (which creates new alleles) and sexual reproduction (which produces new combinations of alleles). Some variants are more suited to the environment of the time than others. These variants will leave more offspring, as described for the hypothetical population (right).

1. Variation through mutation and sexual reproduction:
In a population of brown beetles, mutations independently produce red colouration and 2 spot marking on the wings. The individuals in the population compete for limited resources.



2. Selective predation:
Brown mottled beetles are eaten by birds but red ones are avoided.



3. Change in the genetics of the population:
Red beetles have better survival and fitness and become more numerous with each generation. Brown beetles have poor fitness and become rare.



2. What produces the genetic variation in populations? _____

3. Define evolution: _____

_____

4. Explain how the genetic make-up of a population can change over time: _____

_____

_____

_____

_____

_____

_____

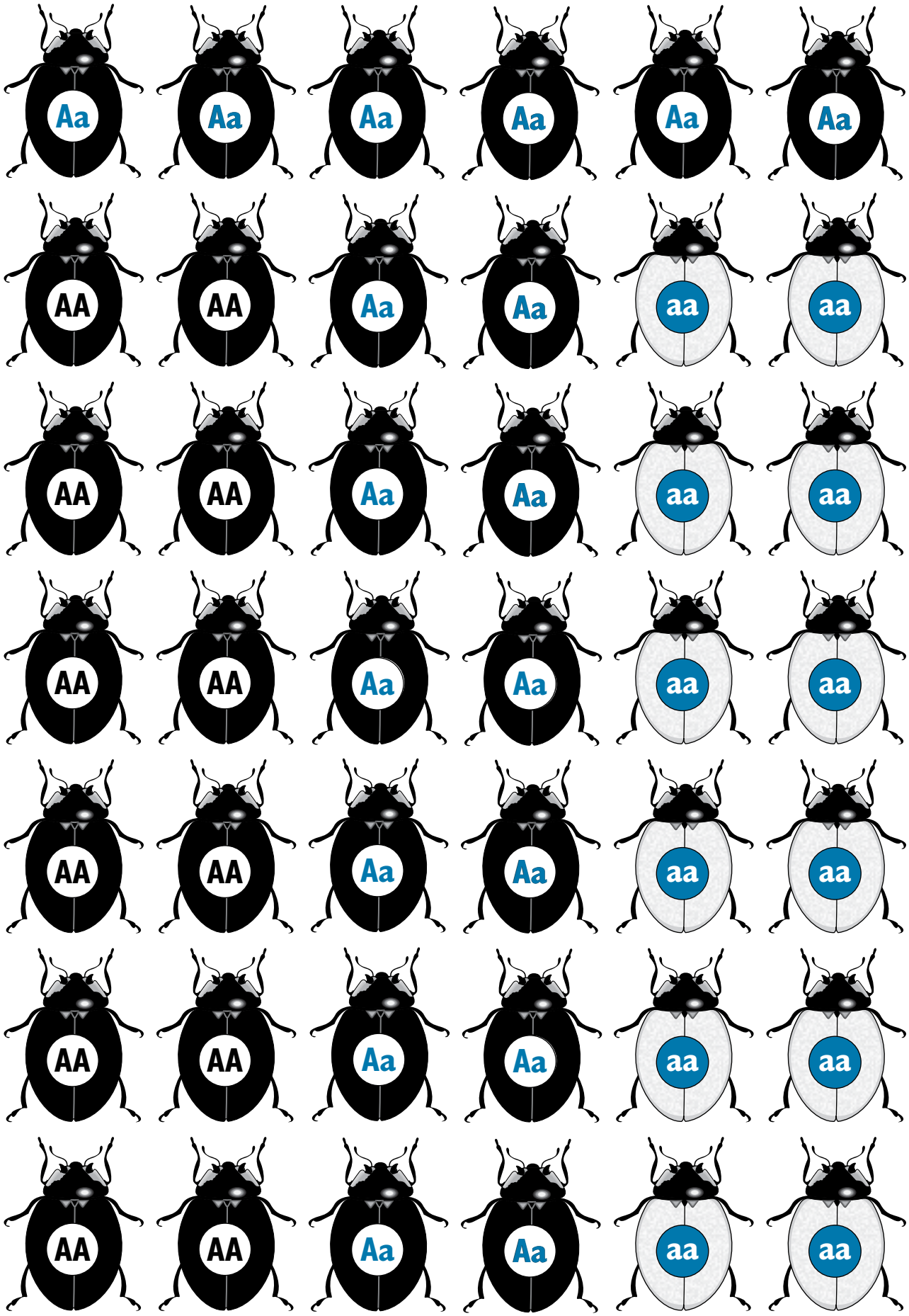
5. Complete the table below by calculating the percentage of beetles in the example above right.

Beetle population	% Brown beetles	% Red beetles	% Red beetles with spots
1			
2			
3			

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205 Gene Pool Exercise

Cut out each of the beetles below and use them to reenact different events within a gene pool as described in this topic pages: *Gene Pools and Evolution, Changes in a Gene Pool, The Founder Effect, Population Bottlenecks, Genetic Drift.*



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206 Gene Pools and Evolution

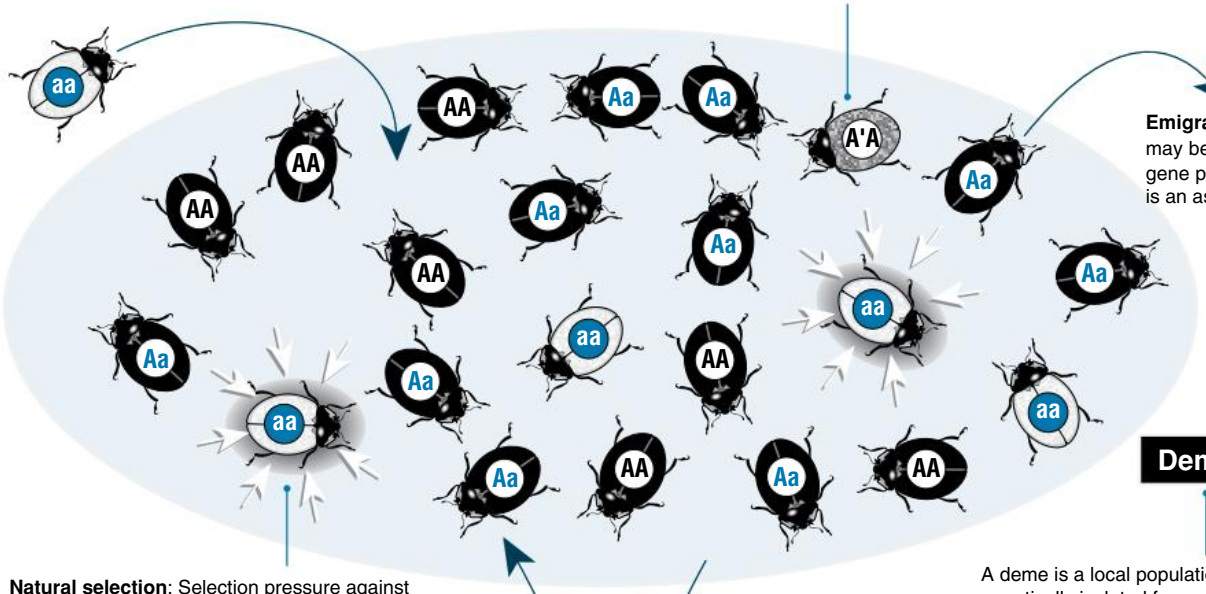
Key Idea: The proportions of alleles in a gene pool can be altered by the processes that increase or decrease variation.

This activity portrays two populations of a beetle species. Each beetle is a 'carrier' of genetic information, represented by the alleles (A and a) for a gene that controls colour and has a dominant/recessive inheritance pattern. There are normally two phenotypes: black and pale. Mutations may create new alleles. Some of the **microevolutionary processes** (natural selection, genetic drift, gene flow, and mutation) that affect the genetic composition (**allele frequencies**) of gene pools are shown below. Simulate the effect of these using the *Gene Pool Exercise*.

Immigration: Populations can gain alleles when they are introduced from other gene pools. Immigration is one aspect of gene flow.

Mutations: Spontaneous mutations can create new alleles. Mutation is very important to evolution, because it is the original source of genetic variation that provides new material for natural selection.

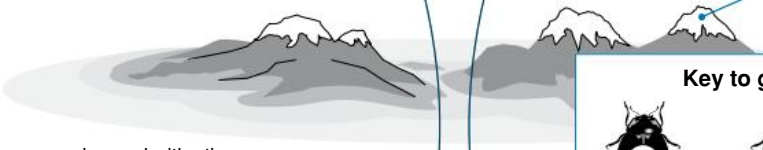
Emigration: Genes may be lost to other gene pools. Emigration is an aspect of gene flow.



Natural selection: Selection pressure against unfavorable allele combinations may reduce survival or reproductive success. Natural selection accumulates and maintains favourable genotypes in a population. It tends to reduce genetic diversity within the gene pool and increase differences between populations.

A deme is a local population that is genetically isolated from other populations of the species.

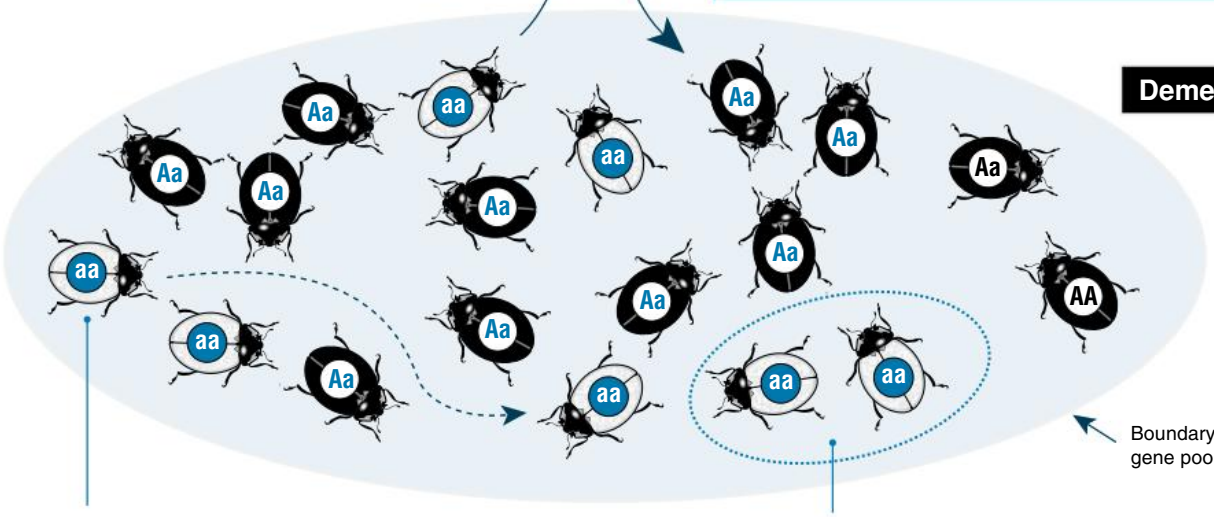
Geographical barriers isolate the gene pool and prevent regular gene flow between populations.



Key to genotypes and phenotypes



Gene flow: Genes are exchanged with other gene pools as individuals move between them. Gene flow is a source of new genetic variation and tends to reduce differences between populations that have accumulated because of natural selection or genetic drift.



Mate choice (non-random mating): Individuals may not select their mate randomly and may seek out particular phenotypes, increasing the frequency of the associated alleles in the population.

Genetic drift: Chance events cause the allele frequencies of small populations to 'drift' (change) randomly from generation to generation. Genetic drift has a relatively greater effect on the genetics of small populations and can be important in their evolution. Small populations may occur as a result of the **founder effect** (where a small number of individuals colonise a new area) or **genetic bottlenecks** (where the population size is dramatically reduced by a catastrophic event) .

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1. For each of the two demes shown on the previous page (treating the mutant in deme 1 as a AA):

   (a) Count up the numbers of **allele types** (**A** and **a**).

   (b) Count up the numbers of **allele combinations** (**AA**, **Aa**, **aa**).

2. Calculate the frequencies as percentages (%) for the allele types and combinations:

Deme 1		Number counted	%
Allele types	A		
	a		
Allele combinations	AA		
	Aa		
	aa		

Deme 2		Number counted	%
Allele types	A		
	a		
Allele combinations	AA		
	Aa		
	aa		

3. One of the fundamental concepts for population genetics is that of **genetic equilibrium**, stated as: *"For a very large, randomly mating population, the proportion of dominant to recessive alleles remains constant from one generation to the next"*. If a gene pool is to remain unchanged, it must satisfy all of the criteria below that favour gene pool stability. Few populations meet all (or any) of these criteria and their genetic makeup must therefore by continually changing. For each of the five factors (a-e) below, state briefly **how** and **why** each would affect the allele frequency in a gene pool:

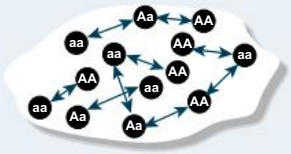
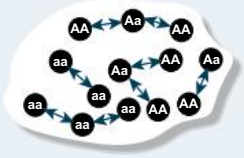
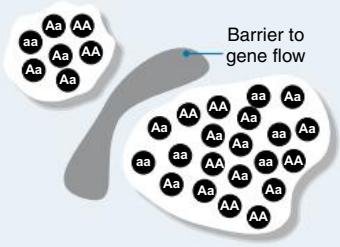
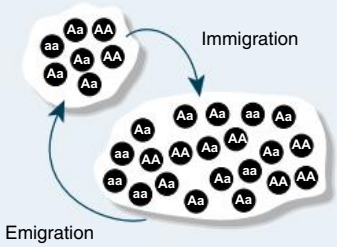
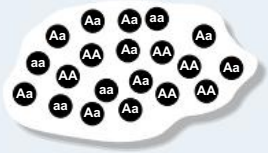
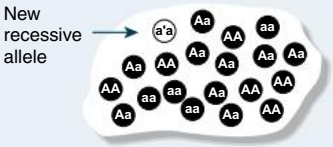
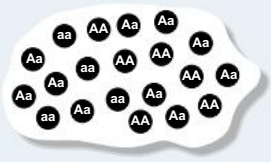
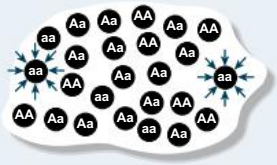
   (a) Population size: _____

   (b) Mate selection: _____

   (c) Gene flow between populations:

   (d) Mutations: _____

   (e) Natural selection: _____

Factors Favouring Gene Pool Stability	Factors Favouring Gene Pool Change
 Large population	 Small population
 Random mating	 Assortative mating
 No gene flow	 Gene flow
 No mutation	 Mutations
 No natural selection	 Natural selection

4. Identify the factors that tend to:

   (a) Increase genetic variation in populations:

   (b) Decrease genetic variation in populations:

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207 Changes in a Gene Pool

Key Idea: Natural selection and migration can alter the allele frequencies in gene pools.

The diagram below shows an hypothetical population of beetles undergoing changes as it is subjected to two 'events'. The three phases represent a progression in time (i.e. the same gene pool, undergoing change). The beetles have two phenotypes (black and pale) determined by the amount of pigment deposited in the cuticle. The gene controlling this character is represented by two alleles **A** and **a**. Your task is to analyse the gene pool as it undergoes changes.

1. For each phase in the gene pool below fill in the tables provided as follows; (some have been done for you):

   (a) Count the number of A and a alleles separately. Enter the count into the top row of the table (left hand columns).
   (b) Count the number of each type of allele combination (AA, Aa and aa) in the gene pool. Enter the count into the top row of the table (right hand columns).
   (c) For each of the above, work out the frequencies as percentages (bottom row of table):

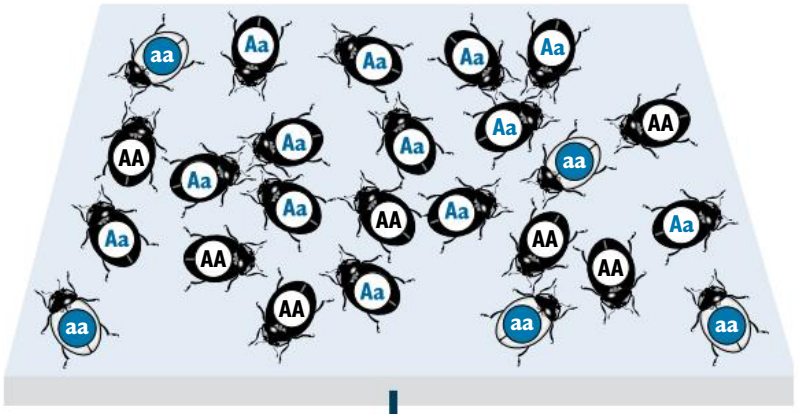
Allele frequency = No. counted alleles ÷ Total no. of alleles x 100

Phase 1: Initial gene pool



	A	a	AA	Aa	aa
No.	27		7		
%	54		28		

Allele types *Allele combinations*

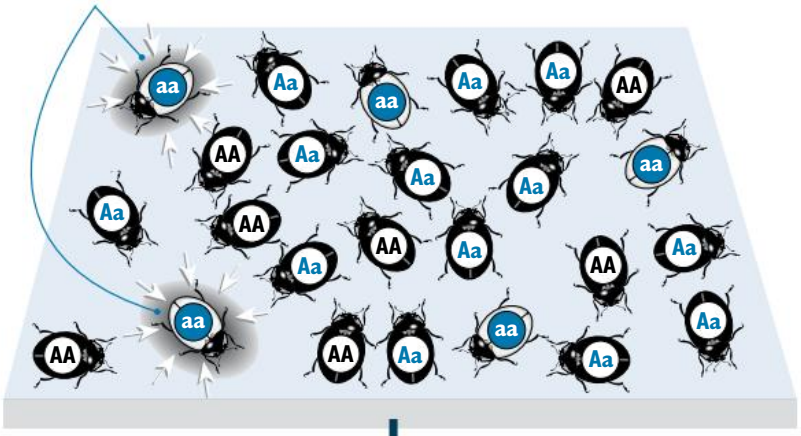


Two pale individuals died. Their alleles are removed from the gene pool.

Phase 2: Natural selection

In the same gene pool at a later time there was a change in the allele frequencies. This was due to the loss of certain allele combinations due to natural selection. Some of those with a genotype of aa were eliminated (poor fitness).

These individuals (surrounded by small white arrows) are not counted for allele frequencies; they are dead!



	A	a	AA	Aa	aa
No.					
%					

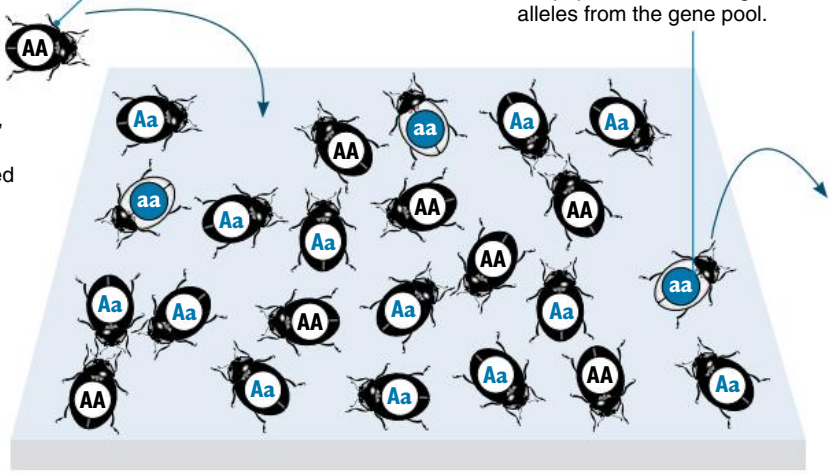
This individual is entering the population and will add its alleles to the gene pool.

This individual is leaving the population, removing its alleles from the gene pool.

Phase 3: Immigration and emigration

This particular kind of beetle exhibits wandering behaviour. The allele frequencies change again due to the introduction and departure of individual beetles, each carrying certain allele combinations.

Individuals coming into the gene pool (AA) are counted for allele frequencies, but those leaving (aa) are not.



	A	a	AA	Aa	aa
No.					
%					

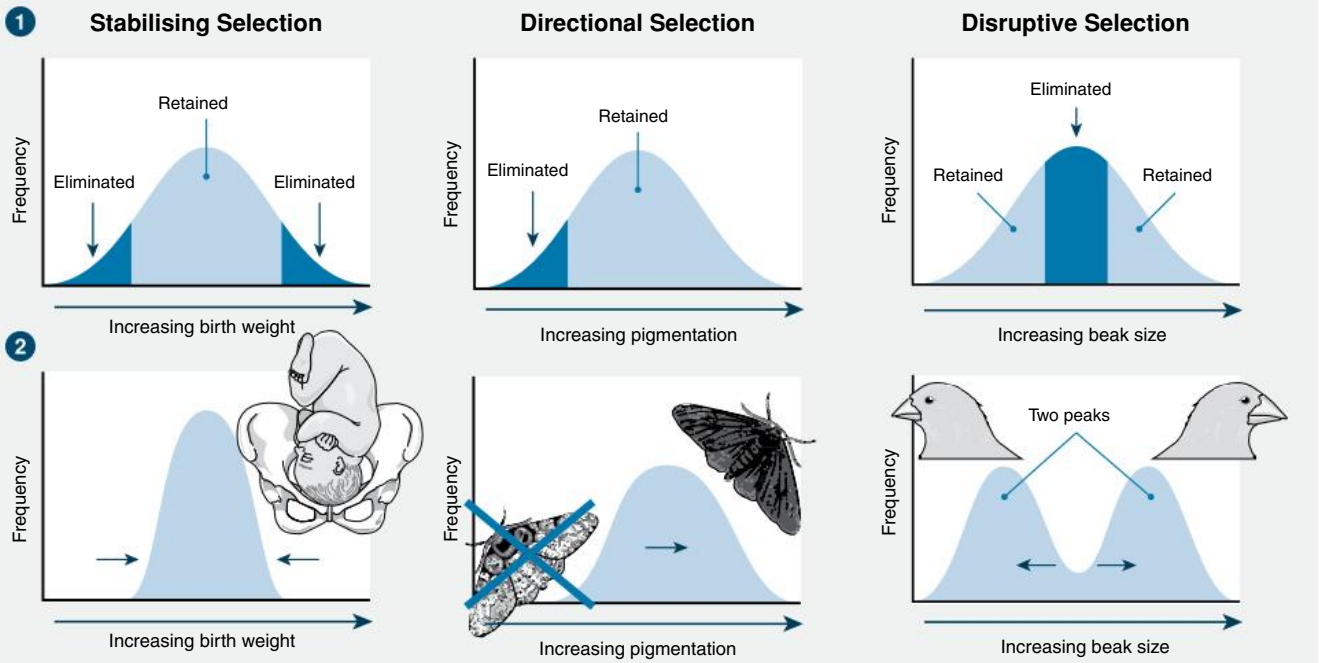
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208 Types of Natural Selection

Key Idea: Natural selection acts on phenotypes and results in the differential survival of some genotypes over others. It is an important cause of change in gene pools.

Natural selection operates on the phenotypes of individuals, produced by their particular combinations of alleles. It results in the differential survival of some genotypes over others. As a result, organisms with phenotypes most suited to the prevailing environment are more likely to survive and breed than those with less suited phenotypes. Favourable phenotypes will become relatively more numerous and than unfavourable phenotypes. Over time, natural selection may lead to a permanent change in the genetic makeup of a population. Natural selection is always linked to phenotypic suitability in the prevailing environment so it is a dynamic process. It may favour existing phenotypes or shift the phenotypic median, as is shown in the diagrams below. The top row of diagrams below represents the population phenotypic spread before selection, and the bottom row the spread afterwards.



Extreme variations are selected against and the middle range (most common) phenotypes are retained in greater numbers. Stabilising selection results in decreased variation for the phenotypic character involved. This type of selection operates most of the time in most populations and acts to prevent divergence of form and function, e.g. birth weight of human infants.

The adaptive phenotype is shifted in one direction and one phenotype is favoured over others. Directional selection was observed in peppered moths in England during the Industrial Revolution. In England's current environment, the selection pressures on the moths are more typically balanced, and the proportions of each form vary regionally.

Disruptive selection favours two phenotypic extremes at the expense of intermediate forms. During a prolonged drought on Santa Cruz Island in the Galápagos, it resulted in a population of ground finches that was bimodal for beak size. Competition for the usual seed sources was so intense that birds able to exploit either small or large seeds were favoured, although intermediate phenotypes remained in low numbers.

1. Explain why fluctuating (as opposed to stable) environments favour disruptive (diversifying) selection:

2. Disruptive selection can be important in the formation of new species:

   (a) Describe the evidence from the ground finches on Santa Cruz Island that provides support for this statement:

   (b) The ground finches on Santa Cruz Island are one interbreeding population with a strongly bimodal distribution for the phenotypic character beak size. Suggest what conditions could lead to the two phenotypic extremes diverging further:

   (c) Predict the consequences of the end of the drought and an increased abundance of medium size seeds as food:

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209 Adaptations and Fitness

Key Idea: Adaptive features enhance an individual's fitness.

An **adaptation** is any heritable trait that suits an organism to its natural role in the environment (its niche). These traits may be structural, physiological, or behavioural. Adaptations promote **fitness**, which is mathematical measure of the contribution an organism makes to the next generation. Adaptations are distinct from properties. Although they may be striking, properties cannot be described as adaptive unless they are shown to be functional in the organism's natural habitat. Genetic adaptation must not be confused with acclimatisation, which refers to an individual's ability to alter its behaviour or physiology in response to its changing environment (e.g. adjustment to altitude in mountaineers). Examples of adaptive features are illustrated below.

Number of Horns in Rhinoceroses

Not all differences between species can be convincingly interpreted as adaptations to particular environments. Rhinoceroses charge rival males and predators, and the horn(s), when combined with the head-down posture, add effectiveness to this behaviour. Horns are obviously adaptive, but it is not clear that the possession of one (Indian rhino) or two (black rhino) horns is necessarily related directly to the environment in which those animals live.


Great Indian rhino


African black rhino

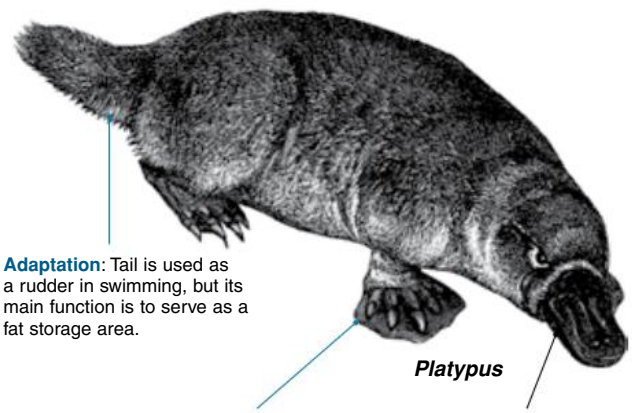
Ear Length in Rabbits and Hares

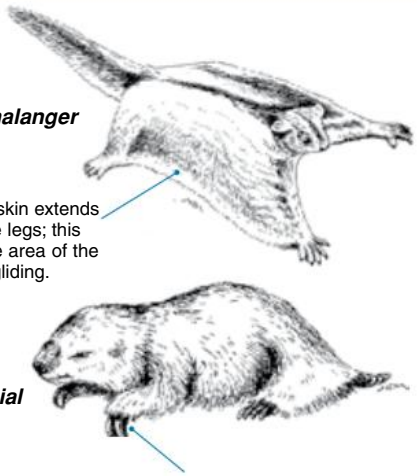
The external ears of many mammals are used as important organs to assist in thermoregulation (controlling loss and gain of body heat). The ears of rabbits and hares native to hot, dry climates, such as the jack rabbit of south-western USA and northern Mexico, are relatively very large. The Arctic hare lives in the tundra zone of Alaska, northern Canada and Greenland, and has ears that are relatively short. This reduction in the size of the extremities (ears, limbs, and noses) is typical of cold adapted species.


Arctic hare: *Lepus arcticus*


Black-tail jackrabbit: *Lepus californicus*

Adaptive Features





1. Distinguish between adaptive features (genetic) and acclimatisation: _____

2. Explain the evolutionary significance of adaptive features: _____

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210 Selection for Skin Colour in Humans

Key Idea: Skin colour is the result of a dynamic balance between two different selection pressures linked to fitness.

Pigmented skin of varying tones is a feature of humans that evolved after early hominins lost the majority of their body hair. The distribution of skin colour globally is not random. People native to equatorial regions have darker skin tones than people from higher latitudes. For many years, biologists postulated that this was because darker skins had evolved to protect against skin cancer. The problem with this explanation was that skin cancer is not tied to evolutionary fitness because it affects post-reproductive individuals and cannot therefore provide a mechanism for selection. More recent in-depth analyses have shown a more complex picture in which selection pressures on skin colour are finely balanced to produce a skin tone that regulates the effects of the sun's ultraviolet radiation on the nutrients vitamin D and folate, both of which are crucial to successful human reproduction, and therefore evolutionary fitness.

Skin Colour in Humans: A Product of Natural Selection

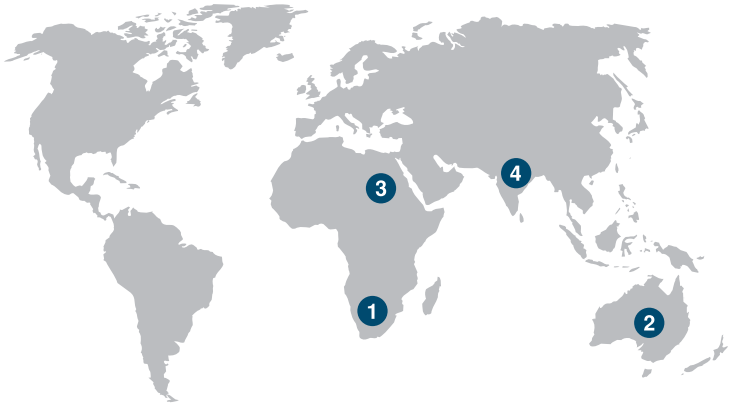


Human skin colour is the result of two opposing selection pressures. Skin pigmentation has evolved to protect against destruction of folate from ultraviolet light, but the skin must also be light enough to receive the light required to synthesise vitamin D. Vitamin D synthesis is a process that begins in the skin and is inhibited by dark pigment. Folate is needed for healthy neural development in humans and a deficiency is associated with fatal neural tube defects. Vitamin D is required for the absorption of calcium from the diet and therefore normal skeletal development.

Women also have a high requirement for calcium during pregnancy and lactation. Populations that live in the tropics receive enough ultraviolet (UV) radiation to synthesise vitamin D all year long. Those that live in northern or southern latitudes do not. In temperate zones, people lack sufficient UV light to make vitamin D for one month of the year. Those nearer the poles lack enough UV light for vitamin D synthesis most of the year (above). Their lighter skins reflect their need to maximise UV absorption (the photos show skin colour in people from different latitudes).

KNOW WEB 210 LINK 208

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Long-term resident **Recent immigrant**

1 Southern Africa: ~ 20-30˚S


Khoisan-Namibia


Zulu: 1000 years ago

2 Australia: ~ 10-35˚S


Aborigine


European: 300 years ago

Rita Willaert-Flickr

3 Banks of the Red Sea: ~ 15-30˚N


Nuba-Sudan


Arab: 2000 years ago

4 India: ~ 10-30˚S


West Bengal


Tamil: ~100 years ago

Aptychus-Flickr

The skin of people who have inhabited particular regions for millennia has adapted to allow sufficient vitamin D production while still protecting folate stores. In the photos above, some of these original inhabitants are illustrated to the left of each pair and compared with the skin tones of more recent immigrants (to the right of each pair, with the number of years since immigration). The numbered locations are on the map.

1. (a) Describe the role of folate in human physiology: _____

   (b) Describe the role of vitamin D in human physiology: _____

2. (a) Early hypotheses to explain skin colour linked pigmentation level only to the degree of protection it gave from UV-induced skin cancer. Explain why this hypothesis was inadequate in accounting for how skin colour evolved:

   _____

   _____

   (b) Explain how the new hypothesis for the evolution of skin colour overcomes these deficiencies: _____

   _____

   _____

   _____

   _____

3. Explain why, in any given geographical region, women tend to have lighter skins (by 3-4% on average) than men:

   _____

   _____

4. The Inuit people of Alaska and northern Canada have a diet rich in vitamin D and their skin colour is darker than predicted on the basis of UV intensity at their latitude. Explain this observation:

   _____

   _____

5. (a) What health problems might be expected for people of African origin now living in northern UK? _____

   _____

   (b) How could these people avoid these problems in their new higher latitude environment? _____

   _____

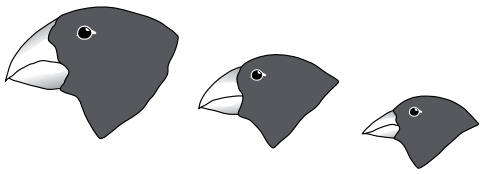
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211 Directional Selection in Darwin's Finches

Key Idea: The effect of directional selection on a population can be verified by making measurements of phenotypic traits. Natural selection acts on the phenotypes of a population. Individuals with phenotypes that increase their fitness produce more offspring, increasing the proportion of the genes corresponding to that phenotype in the next generation. Many population studies have shown natural selection can cause phenotypic changes in a population relatively quickly.

The finches on the Galápagos island (Darwin's finches) are famous in that they are commonly used as examples of how evolution produces new species. In this activity you will analyse data from the measurement of beaks depths of the medium ground finch (*Geospiza fortis*) on the island of Daphne Major near the centre of the Galápagos Islands. The measurements were taken in 1976 before a major drought hit the island and in 1978 after the drought (survivors and survivors' offspring).



Beak depth (mm)	No. 1976 birds	No. 1978 survivors
7.30-7.79	1	0
7.80-8.29	12	1
8.30-8.79	30	3
8.80-9.29	47	3
9.30-9.79	45	6
9.80-10.29	40	9
10.30-10.79	25	10
10.80-11.29	3	1
11.30+	0	0

Beak depth of offspring (mm)	Number of birds
7.30-7.79	2
7.80-8.29	2
8.30-8.79	5
8.80-9.29	21
9.30-9.79	34
9.80-10.29	37
10.30-10.79	19
10-80-11.29	15
11.30+	2

1. Use the data above to draw two separate sets of histograms:

   (a) On the left hand grid draw side-by-side histograms for the number of 1976 birds per beak depth and the number of 1978 survivors per beak depth.

   (b) On the right hand grid draw a histogram of the beak depths of the offspring of the 1978 survivors.

2. (a) Mark the approximate mean beak depth on the graphs of the 1976 beak depths and the 1978 offspring.

   (b) How much has the average moved from 1976 to 1978? _____

   (c) Is beak depth heritable? What does this mean for the process of natural selection in the finches?

   _____

   _____

3. The 1976 drought resulted in plants dying back and not producing seed. Based on the graphs, what can you say about competition between the birds for the remaining seeds, i.e. in what order were the seeds probably used up?

   _____

   _____

   _____

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212 Disruptive Selection in Darwin's Finches

Key Idea: Disruptive selection in the finch *Geospiza fortis* produces a bimodal distribution for beak size.

The Galápagos Islands, 970 km west of Ecuador, are home to the finch species *Geospiza fortis*. A study during a prolonged drought on Santa Cruz Island showed how **disruptive selection** can change the distribution of genotypes in a population. During the drought, large and small seeds were more abundant than the preferred intermediate seed size.

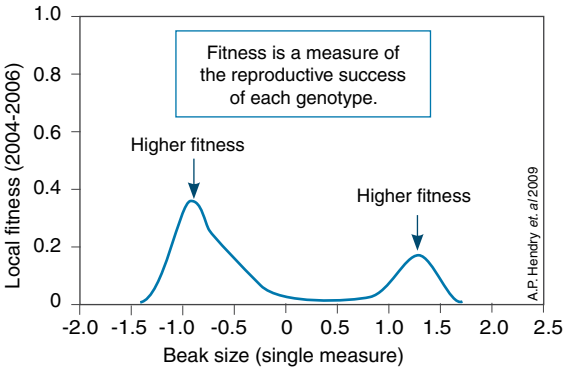
Beak sizes of *G. fortis* were measured over a three year period (2004-2006), at the start and end of each year. At the start of the year, individuals were captured, banded, and their beaks were measured.

The presence or absence of banded individuals was recorded at the end of the year when the birds were recaptured. Recaptured individuals had their beaks measured.

The proportion of banded individuals in the population at the end of the year gave a measure of fitness. Absent individuals were presumed dead (fitness = 0).

Fitness related to beak size showed a bimodal distribution (left) typical of disruptive selection.

Beak size vs fitness in *Geospiza fortis*

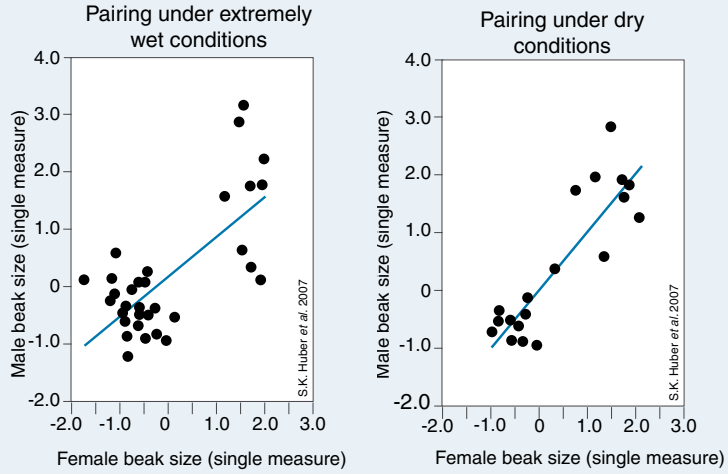


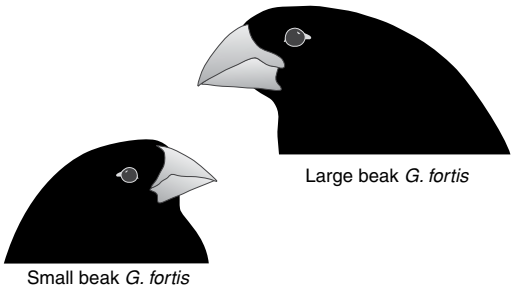
*Fitness showed a **bimodal distribution** (arrowed) being highest for smaller and larger beak sizes.*

Measurements of the beak length, width, and depth were combined into one **single measure**.



Beak Size Pairing in *Geospiza fortis*





A 2007 study found that breeding pairs of birds had similar beak sizes. Male and females with small beaks tended to breed together, and males and females with large beaks tended to breed together. Mate selection maintained the biomodal distribution in the population during extremely wet conditions. If beak size wasn't a factor in mate selection, the beak size would even out.

1. (a) How did the drought affect seed size on Santa Cruz Island? _____

_____

(b) How did the change in seed size during the drought create a selection pressure for changes in beak size?

_____

_____

_____

2. How does beak size relate to fitness (differential reproductive success) in *G. fortis*? _____

_____

_____

_____

3. (a) Is mate selection in *G. fortis* random / non-random? (delete one)

(b) Give reasons for your answer: _____

_____

_____

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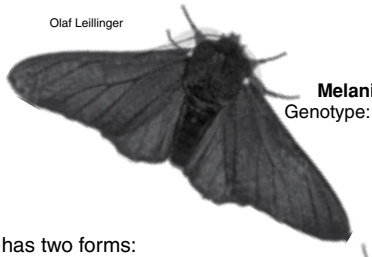
213 Directional Selection in Moths

Key Idea: Directional selection pressures on the peppered moth during the Industrial Revolution shifted the common phenotype from the grey form to the melanic (dark) form.

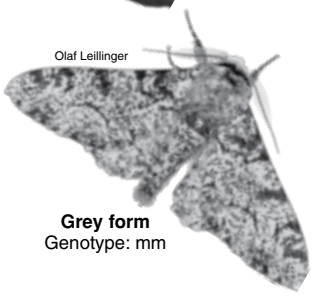
Natural selection may act on the frequencies of phenotypes (and hence genotypes) in populations in one of three different ways (through stabilising, directional, or disruptive selection).

Colour change in the **peppered moth** (*Biston betularia*) during the Industrial Revolution is often used to show **directional selection** in a polymorphic population (polymorphic means having two or more forms). Intensive coal burning during this time caused trees to become dark with soot, and the dark form (morph) of peppered moth became dominant.

The gene controlling colour in the peppered moth, is located on a single locus. The allele for the melanic (dark) form (**M**) is dominant over the allele for the grey (light) form (**m**).


Olaf Leillinger

Melanic form
Genotype: MM or Mm


Olaf Leillinger

Grey form
Genotype: mm

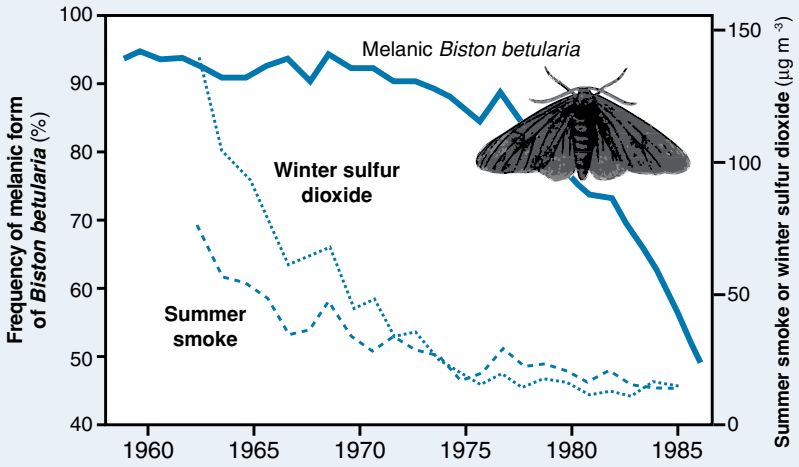
The peppered moth, *Biston betularia*, has two forms: a grey mottled form, and a dark melanic form. During the Industrial Revolution, the relative abundance of the two forms changed to favour the dark form. The change was thought to be the result of selective predation by birds. It was proposed that the grey form was more visible to birds in industrial areas where the trees were dark. As a result, birds preyed upon them more often, resulting in higher numbers of the dark form surviving.



Museum collections of the peppered moth over the last 150 years show a marked change in the frequency of the melanic form (above right). Moths collected in 1850, prior to the major onset of the Industrial Revolution in England, were mostly the grey form (above left). Fifty years later the frequency of the darker melanic forms had increased.

In the 1940s and 1950s, coal burning was still at intense levels around the industrial centres of Manchester and Liverpool. During this time, the melanic form of the moth was still very dominant. In the rural areas further south and west of these industrial centres, the occurrence of the grey form increased dramatically. With the decline of coal burning factories and the introduction of the Clean Air Act in cities, air quality improved between 1960 and 1980. Sulfur dioxide and smoke levels dropped to a fraction of their previous levels. This coincided with a sharp fall in the relative numbers of melanic moths (right).

Frequency of melanic peppered moth related to reduced air pollution



1. The populations of peppered moth in England have undergone changes in the frequency of an obvious phenotypic character over the last 150 years. What is the phenotypic character?

2. Describe how the selection pressure on the grey form has changed with change in environment over the last 150 years:

3. Describe the relationship between allele frequency and phenotype frequency:

4. The level of pollution dropped around Manchester and Liverpool between 1960 and 1985. How did the frequency of the darker melanic form change during this period?

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214 Selection for Human Birth Weight

Key Idea: Stabilising selection operates to keep human birth weight within relatively narrow constraints.

Selection pressures operate on populations in such a way as to reduce mortality. For humans, giving birth is a special, but often traumatic, event. In a study of human birth weights it is possible to observe the effect of selection pressures operating to constrain human birth weight within certain limits. This is a good example of **stabilising selection**. This activity explores the selection pressures acting on the birth weight of human babies. Carry out the steps below:

Step 1: Collect the birth weights from 100 birth notices from your local newspaper (or 50 if you are having difficulty getting enough; this should involve looking back through the last 2-3 weeks of birth notices). If you cannot obtain birth weights in your local newspaper, a set of 100 sample birth weights is provided in the Model Answers booklet.

Step 2: Group the weights into each of the 12 weight classes (of 0.5 kg increments). Determine what percentage (of the total sample) fall into each weight class (e.g. 17 babies weigh 2.5-3.0 kg out of the 100 sampled = 17%)

Step 3: Graph these in the form of a histogram for the 12 weight classes (use the graphing grid provided right). Be sure to use the scale provided on the left vertical (y) axis.

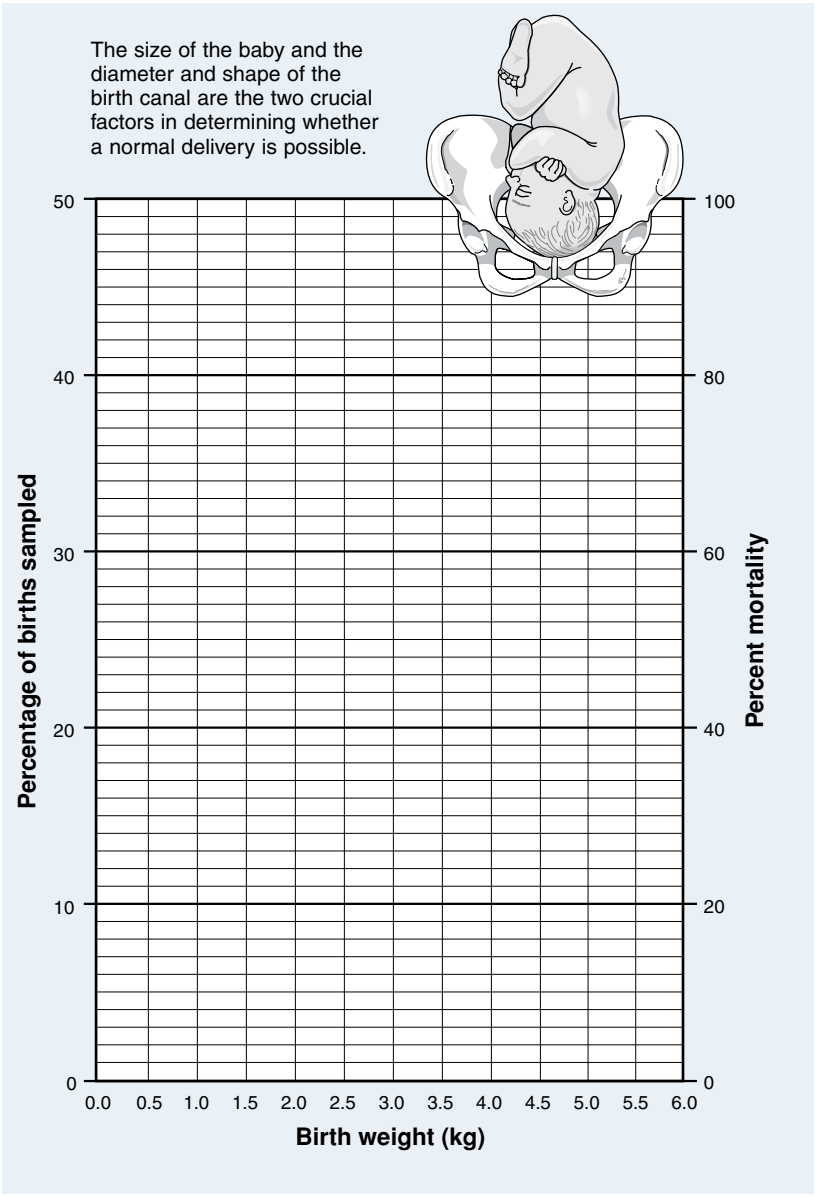
Step 4: Create a second graph by plotting percentage mortality of newborn babies in relation to their birth weight. Use the scale on the right y axis and data provided (below).

Step 5: Draw a line of 'best fit' through these points.

Mortality of newborn babies related to birth weight

Weight (kg)	Mortality (%)
1.0	80
1.5	30
2.0	12
2.5	4
3.0	3
3.5	2
4.0	3
4.5	7
5.0	15

Source: Biology: The Unity & Diversity of Life (4th ed), by Starr and Taggart



1. Describe the shape of the histogram for birth weights: _____

2. What is the optimum birth weight in terms of the lowest newborn mortality? _____

3. Describe the relationship between newborn mortality and birth weight: _____

_____

4. Describe the selection pressures that are operating to control the range of birth weight: _____

_____

_____

5. How might have modern medical intervention during pregnancy and childbirth altered these selection pressures?

_____

_____

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215 Heterozygous Advantage

Key Idea: Heterozygous advantage is a phenomenon in which the heterozygote has a greater fitness in the prevailing environment than either of the homozygotes.

Natural selection operates on phenotypes (and therefore their genotypes) in the prevailing environment. For some phenotypic conditions controlled by a single gene with two alleles, a heterozygote (an individual with two different alleles for a gene) may have a higher fitness than either of the homozygous conditions. This situation is called **heterozygous advantage**. In the case of the sickle cell allele outlined below, susceptibility to malaria is high in the homozygous dominant condition, but lower in the heterozygous condition. Consequently, the heterozygote has a higher fitness in malaria-prone regions. Heterozygous advantage can result in the stable coexistence of different phenotypes in a population (a state called **balanced polymorphism**) and can account for the persistence of detrimental alleles. The maintenance of the sickle cell mutation in malaria-prone regions is one of the few well documented examples in which the evidence for heterozygous advantage is conclusive.

The Sickle Cell Allele (HbS)

Sickle cell disease is caused by a mutation in a gene that directs the production of the human blood protein called haemoglobin. Genetic analyses show that the mutation arose spontaneously in different regions. The mutant allele is known as **Hb^S** and produces a form of haemoglobin that differs from the normal form by just one amino acid in the β-chain. This small change however causes a cascade of physiological problems in people with the allele. The red blood cells containing mutated haemoglobin alter their shape to become irregular and spiky: having a **sickle cell** appearance.

Sickle cells have a tendency to clump together and work less efficiently. In people with just one sickle cell allele plus a normal allele (the heterozygote condition **Hb^SHb**), there is a mixture of both red blood cell types and they are said to have the sickle cell trait. They are generally unaffected by the disease. People with two Hb^S genes (**Hb^SHb^S**) suffer severe illness and often die prematurely. HbS is therefore considered a **lethal gene**.

Heterozygous Advantage in Malarial Regions

Falciparum malaria is widely distributed throughout central Africa, the Mediterranean, Middle East, and tropical and semi-tropical Asia (Fig. 1). It is transmitted by the *Anopheles* mosquito, which spreads the protozoan *Plasmodium falciparum* from person to person as it feeds on blood.

SYMPTOMS: These appear 1-2 weeks after being bitten, and include headache, shaking, chills, and fever. Falciparum malaria is more severe than other forms of malaria, with high fever, convulsions, and coma. Death can occur within days of the first symptoms appearing.

THE PARADOX: The Hb^S allele offers considerable protection against malaria. Sickle cells have low potassium levels, which causes *Plasmodium* parasites inside these cells to die. Those with a normal phenotype are very susceptible to malaria, but heterozygotes (Hb^SHb) are much less so. This situation, called **heterozygous advantage**, has resulted in the Hb^S allele being present in moderately high frequencies in parts of Africa and Asia despite its harmful effects (Fig. 2). This is a special case of balanced polymorphism, called a **balanced lethal system** because neither of the homozygotes produces a phenotype that survives, but the heterozygote is viable.

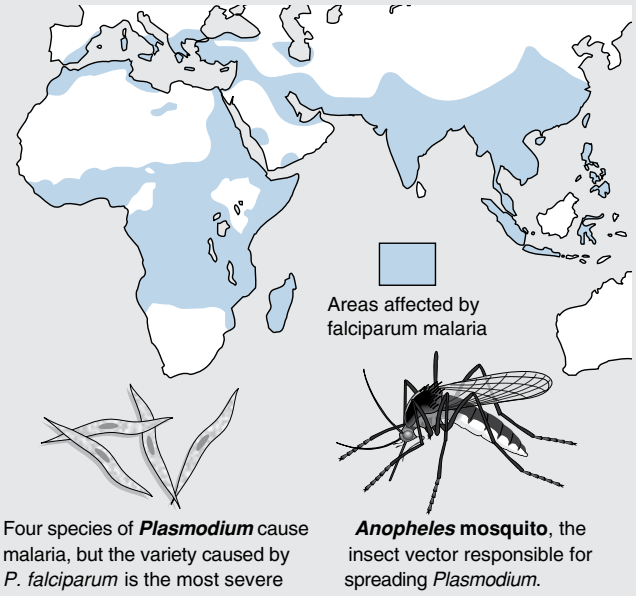


Fig. 1: Incidence of falciparum malaria

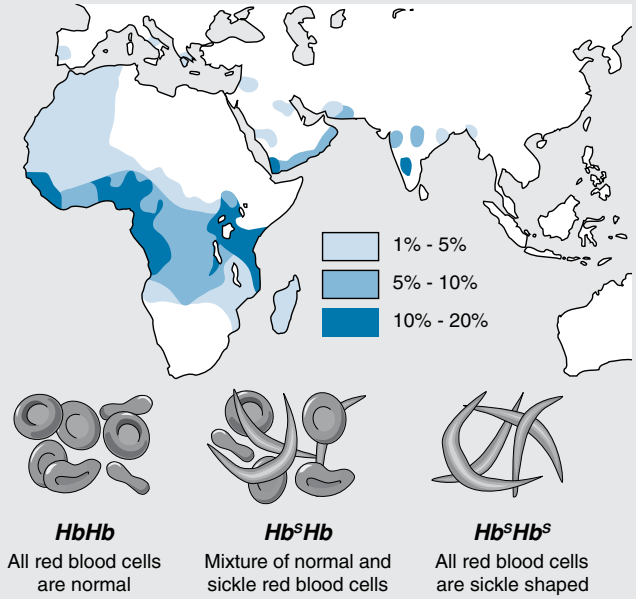


Fig. 2: Frequency of the sickle cell allele

1. Why do carriers of the Hb^s allele have an advantage in malaria-prone regions? _____

_____

_____

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2. With respect to the sickle cell allele, explain how **heterozygous advantage** can lead to **balanced polymorphism**:

3. (a) Describe the distribution of malaria throughout the world (Fig. 1):

   (b) What do you notice about its distribution compared to the frequency of the Hb^S allele (Fig. 2)?

Figure 3 shows the general distributions of various haemoglobin disorders that all produce abnormal red blood cells to some degree.

Hb^E is a mutation that appears to have arisen about 5000 years ago and is caused by a change in the 26th amino acid in the β-chain from glutamic acid to lysine.

The Hb^S mutation changes the 6th amino acid from glutamic acid to valine.

Hb^C is a mutation that occurs in the same position as Hb^S, but the mutation produces the amino acid lysine instead of valine.

Both Hb^E and Hb^C heterozygotes show virtually no (and certainly much less than Hb^S) symptoms of anaemia (reduced haemoglobin levels).

Thalassaemia is a disease in which gene mutations result in the lowered production of haemoglobin and red blood cells. The effects can be very severe.

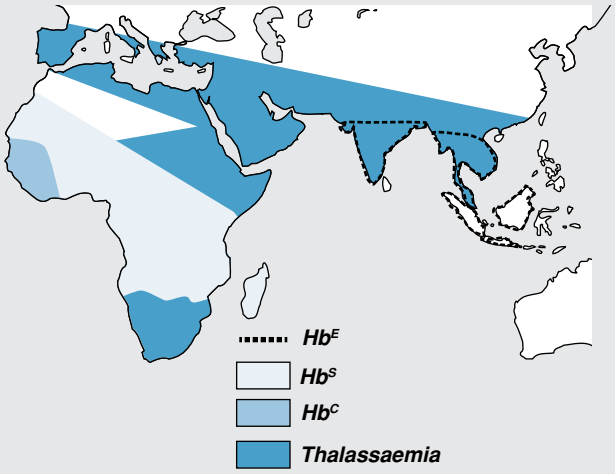


Fig. 3: Distribution of abnormal blood conditions

4. Account for the distribution of red blood cell abnormalities, explaining why these abnormalities persist:

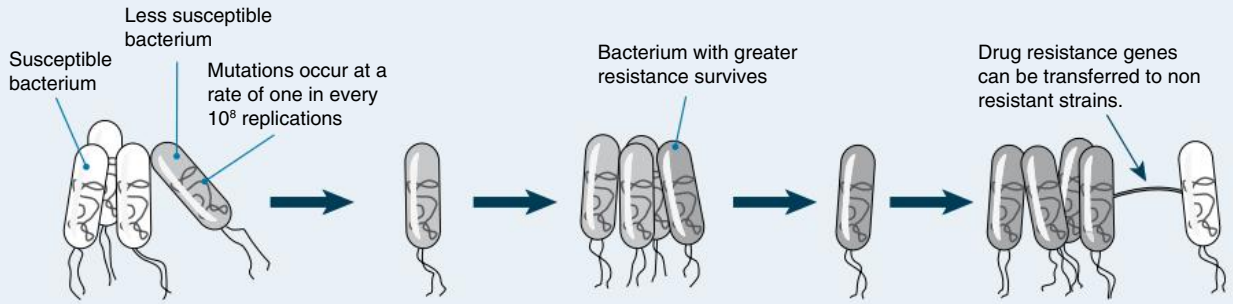
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216 The Evolution of Antibiotic Resistance

Key Idea: Current widespread use of antibiotics has created a selective environment for the proliferation of antibiotic resistance in bacterial populations.

Antibiotic resistance arises when a genetic change allows bacteria to tolerate levels of antibiotic that would normally inhibit growth. This resistance may arise spontaneously, through mutation or copying error, or by transfer of genetic material between microbes. Genomic analyses from 30,000 year old permafrost sediments show that the genes for antibiotic resistance are not new. They have long been present in the bacterial genome, predating the modern selective pressure of antibiotic use. In the current selective environment, these genes have proliferated and antibiotic resistance has spread. For example, methicillin resistant strains of *Staphylococcus aureus* (MRSA) have acquired genes for resistance to all penicillins. Such strains are called superbugs.

The Evolution of Drug Resistance in Bacteria

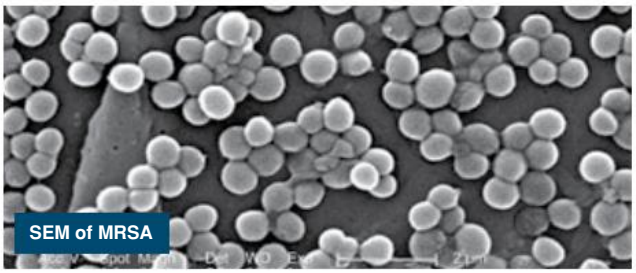


Any population, including bacterial populations, includes variants with unusual traits, in this case reduced sensitivity to an antibiotic. These variants arise as a result of mutations in the bacterial chromosome.

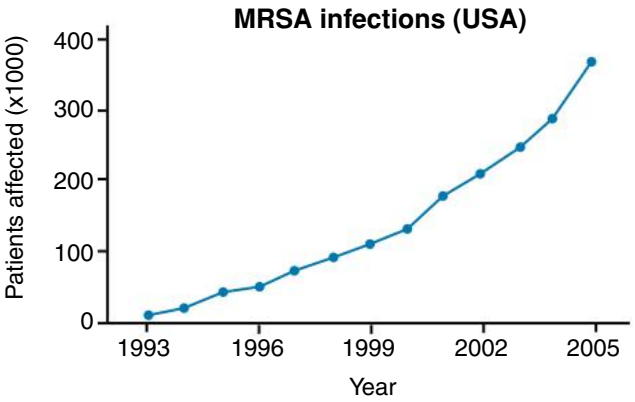
When a person takes an antibiotic, only the most susceptible bacteria will die. The more resistant cells remain alive and continue dividing. Note that the antibiotic does not create the resistance; it provides the environment in which selection for resistance can take place.

If the amount of antibiotic delivered is too low, or the course of antibiotics is not completed, a population of resistant bacteria develops. Within this population too, there will be variation in susceptibility. Some will survive higher antibiotic levels than others.

A highly resistant population has evolved. The resistant cells can exchange genetic material with other bacteria (via horizontal gene transmission), passing on the genes for resistance. The antibiotic initially used against this bacterial strain will now be ineffective.



Staphylococcus aureus is a common bacterium responsible for several minor skin infections in humans. MRSA is a strain that has evolved resistance to penicillin and related antibiotics. MRSA is troublesome in hospital-associated infections because patients with open wounds, invasive devices (e.g. catheters), or poor immunity are at greater risk for infection than the general public.



1. What does **antibiotic resistance** mean? _____

_____

2. (a) How does antibiotic resistance arise in a bacterial population? _____

_____

(b) Describe two ways in which antibiotic resistance can become widespread: _____

_____

_____

3. With reference to MRSA, describe the implications to humans of widespread antibiotic resistance:

_____

_____

_____

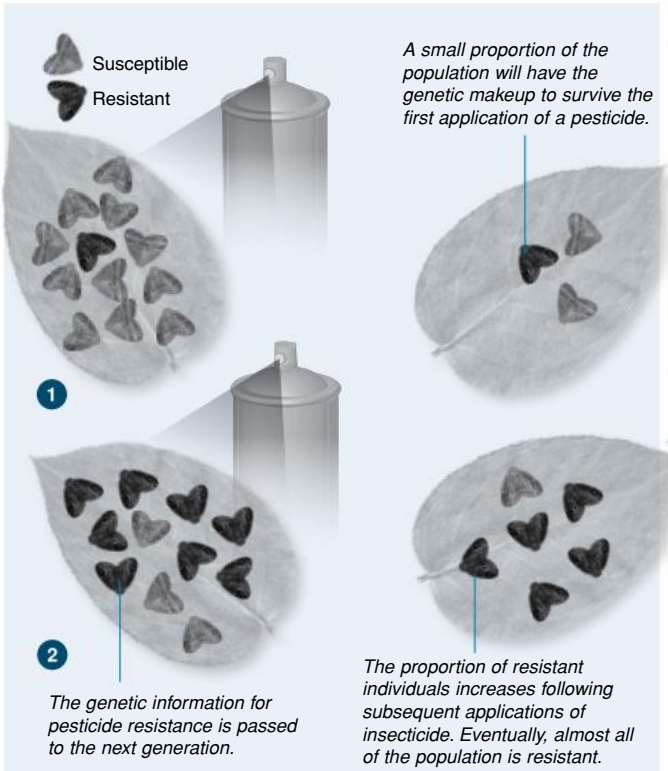
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217 Insecticide Resistance

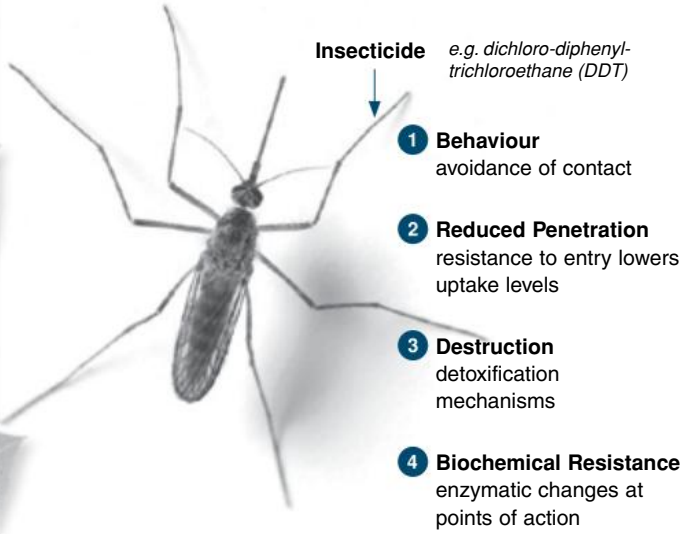
Key Idea: Insect resistance to insecticide is increasing as a result of ineffectual initial applications of insecticide that only kill the most susceptible insects, leaving the more resistant ones to form a new, more resistant population.

Insecticides are pesticides used to control pest insects. They have been used for hundreds of years, but their use has increased since synthetic insecticides were first developed in the 1940s. When **insecticide resistance** develops, the control agent will no longer control the target species. Resistance can arise through behavioural, anatomical, biochemical, and physiological mechanisms, but the underlying process is a form of **natural selection**, in which the most resistant organisms survive to pass on their genes to their offspring. To combat increasing resistance, higher doses of more potent pesticides are sometimes used. This drives the selection process, so that increasingly higher dose rates are required to combat rising resistance. This phenomenon is made worse by the development of multiple resistance in some pest species. Insecticides are widely used, so the development of resistance has serious environmental and economic consequences.



The Development of Resistance

The application of an insecticide can act as a potent selection pressure for resistance in pest insects. The insecticide acts as a selective agent, and only individuals with greater natural resistance survive the application to pass on their genes to the next generation. These genes (or combination of genes) may spread through all subsequent populations.



Mosquitoes are vectors for malaria in humans. In 1955, a global malaria eradication programme was established to eliminate malaria by residual low level spraying of DDT. The programme was effective at first, but resistance soon developed in many mosquito populations. It was eventually abandoned in 1969.

Mechanisms of Resistance in Insect Pests

Insecticide resistance in insects can arise through a combination of mechanisms. (1) Increased sensitivity to an insecticide will cause the pest to avoid a treated area. (2) Certain genes (e.g. the *PEN* gene) confer stronger physical barriers, decreasing the rate at which the chemical penetrates the cuticle. (3) Detoxification by enzymes within the insect's body can render the pesticide harmless, and (4) structural changes to the target enzymes make the pesticide ineffective. No single mechanism provides total immunity, but together they transform the effect from potentially lethal to insignificant.

1. Give two reasons why widespread insecticide resistance can develop very rapidly in insect populations:

   (a) _____

   (b) _____

2. Explain how repeated insecticide applications act as a selective agent for evolutionary change in insect populations:

   _____

   _____

3. With reference to synthetic insecticides, discuss the implications of insecticide resistance to human populations:

   _____

   _____

   _____

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LINK 204 KNOW

218 Antigenic Variability in Pathogens

Key Idea: Rapid evolution in *Influenzavirus* is indicated by the genetically encoded changes to membrane surface proteins. These changes are adaptations to its human host.

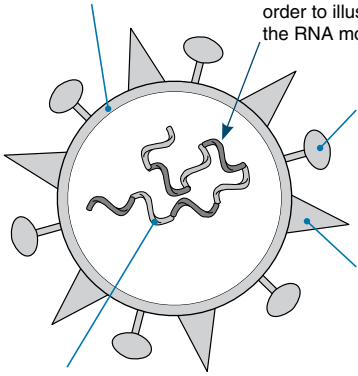
Influenza (flu) is a disease of the upper respiratory tract caused by the viral genus *Influenzavirus*. Three types of *Influenzavirus* (A, B, and C) affect humans. The most common and most virulent of these is *Influenzavirus* A, (below left). Influenza viruses are constantly undergoing genetic changes by either **antigenic drift** or **antigenic shift** (below right). These genetic changes result in changes to the proteins presented on the virus's surface. The changes to the proteins prevent the human immune system from detecting the virus easily, and allow the virus to reinfect people who may already have had the flu.

Structure of *Influenzavirus*

Viral strains are identified by the variation in their H and N surface antigens. Viruses are able to combine and readily rearrange their RNA segments, which alters the protein composition of their H and N glycoprotein spikes.

The *influenzavirus* is surrounded by an **envelope** containing protein and lipids.

The genetic material is actually closely surrounded by protein capsomeres (these have been omitted here and below right in order to illustrate the changes in the RNA more clearly).



The **neuraminidase (N) spikes** help the virus to detach from the cell after infection.

Hemagglutinin (H) spikes allow the virus to recognise and attach to cells before attacking them.

The viral genome is contained on **eight RNA segments**, which enables the exchange of genes between different viral strains.

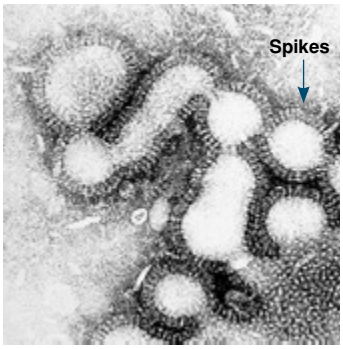


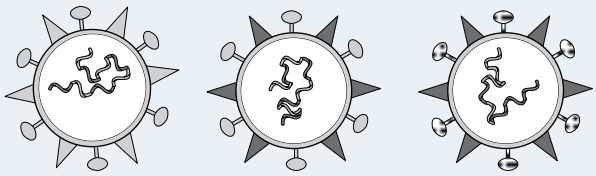
Photo right: *Electron micrograph of Influenzavirus showing the glycoprotein spikes projecting from the viral envelope*

Change in *Influenzavirus*

Antigenic drifts are small changes in the virus which happen continually over time. Such changes mean that the influenza vaccine must be adjusted each year to include the most recently circulating influenza viruses.

Antigenic shift occurs when two or more different viral strains (or different viruses) combine to form a new subtype. The changes are large and sudden and most people lack immunity to the new subtype. New influenza viruses arising from antigenic shift have caused influenza pandemics that have killed millions people over the last century. *Influenzavirus* A is dangerous to human health because it is capable of antigenic shift.

***Influenzavirus* subtypes in humans**



H1N1 ***H1N2*** ***H3N2***



CDC

Influenza vaccination is the primary method for preventing influenza and is 75% effective. The ability of the virus to recombine its RNA enables it to change each year, so that different strains occur each 'flu' season.

The 'flu' vaccination is updated annually to incorporate the antigenic properties of currently circulating strains. Three strains are chosen for each year's vaccination. Selection is based on estimates of which strains will be predominant in the following year.

1. The *Influenzavirus* is able to mutate readily and alter the composition of H and N spikes on its surface.

   (a) Why can the virus mutate so rapidly? _____

   _____

   _____

   (b) How does this affect the ability of the immune system to recognise and respond to the virus? _____

   _____

   _____

   _____

2. Discuss why a virus capable of antigenic shift is more dangerous to humans than a virus undergoing antigenic drift:

   _____

   _____

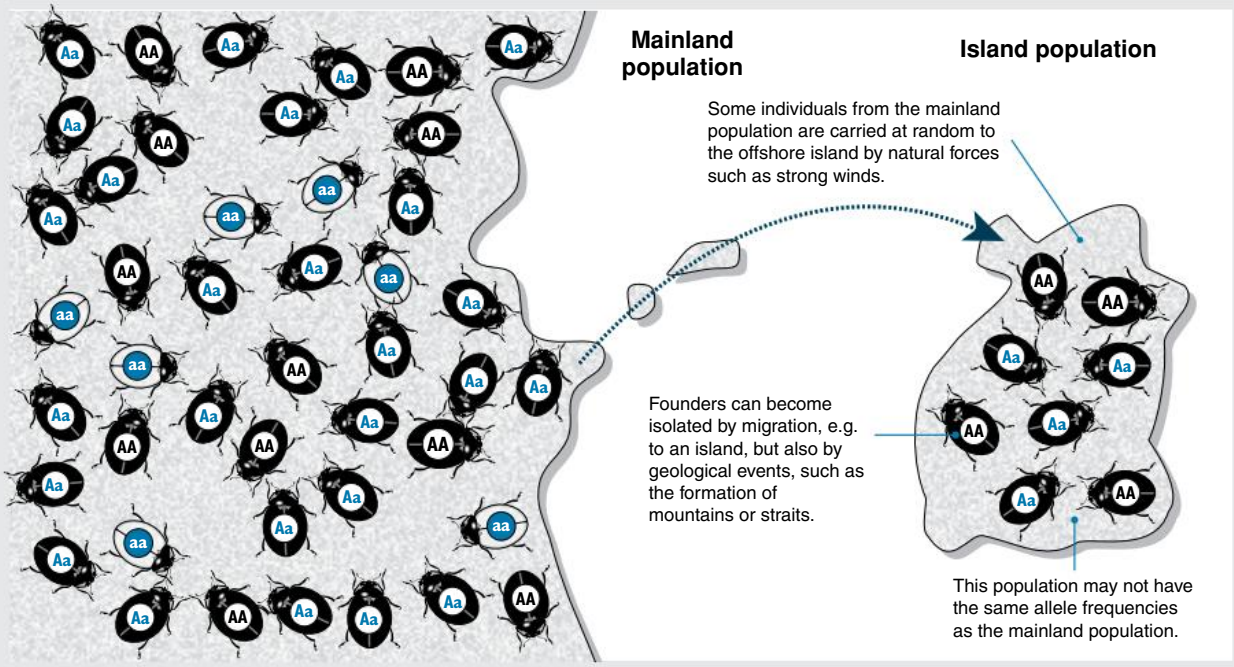
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219 The Founder Effect

Key Idea: The founder effect can result in differences in allele frequencies between a parent and founder populations.

If a small number of individuals from a large population becomes isolated from their original parent population, their sample of alleles is unlikely to represent the allele proportions of the parent population. This phenomenon is called the **founder effect** and it can result in the colonising (founder) population evolving in a different direction to the parent population. This is particularly the case if the founder population is subjected to different selection pressures in a new environment and if the population is missing alleles that are present in the parent population.



Mainland Population

	Allele frequencies		Phenotype frequencies	
	Actual numbers	Calculate %	Black	Pale
Allele A				
Allele a				
Total				

Colonising Island Population

	Allele frequencies		Phenotype frequencies	
	Actual numbers	Calculate %	Black	Pale
Allele A				
Allele a				
Total				

1. Compare the mainland population to the population which ended up on the island (use the spaces in the tables above):
   (a) Count the **phenotype** numbers for the two populations (i.e. the number of black and pale beetles).
   (b) Count the **allele** numbers for the two populations: the number of dominant alleles (A) and recessive alleles (a). Calculate these as a percentage of the total number of alleles for each population.

2. How are the allele frequencies of the two populations different? _____

3. Describe some possible ways in which various types of organism can be **carried** to an offshore island:
   (a) Plants: _____
   (b) Land animals: _____
   (c) Non-marine birds: _____

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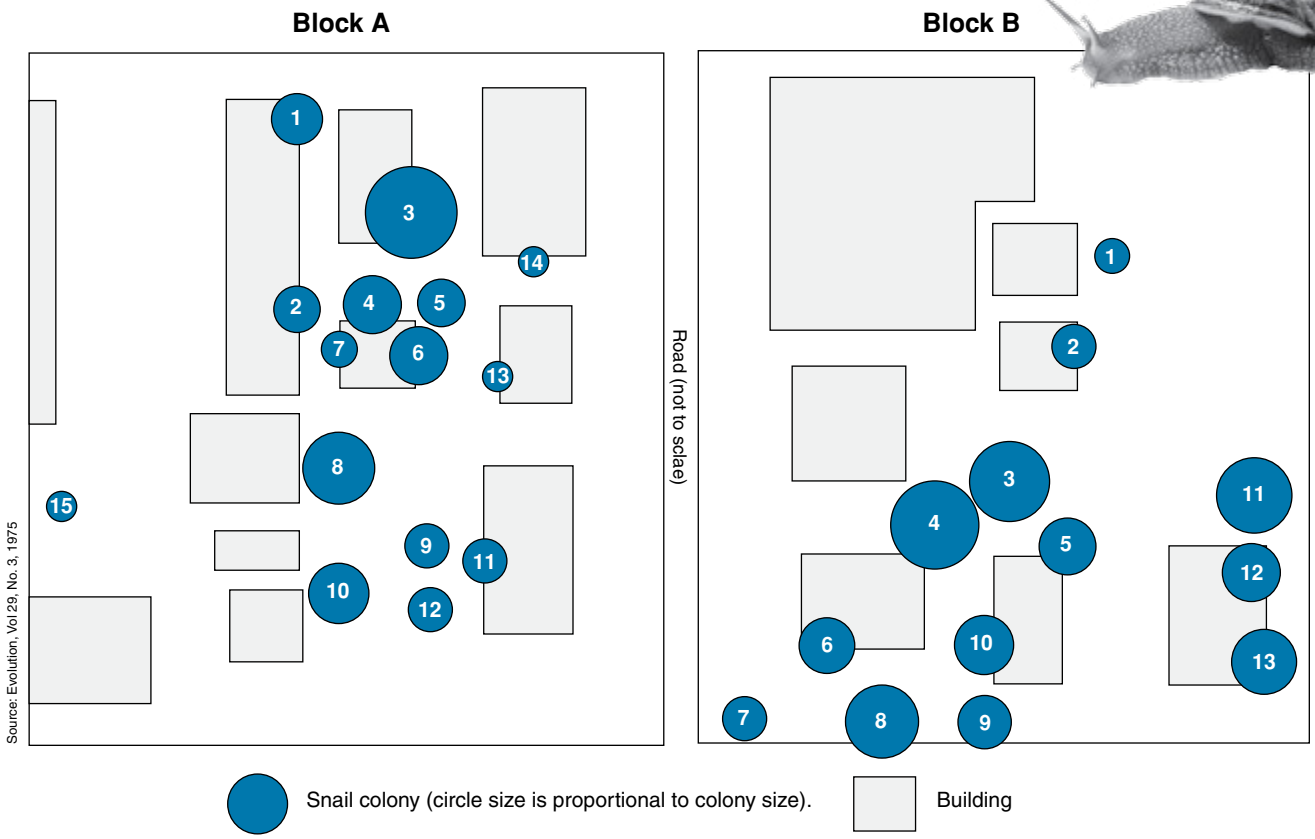
LINK 229 LINK 221 LINK 205 WEB 219 DATA

Microgeographic Isolation in Garden Snails

The European garden snail (*Cornu aspersum*, formerly *Helix aspersa*) is widely distributed throughout the world, both naturally and by human introduction. However because of its relatively slow locomotion and need for moist environments it can be limited in its habitat and this can lead to regional variation. The study below illustrates an investigation carried out on two snail populations in the city of Bryan, Texas. The snail populations covered two adjacent city blocks surrounded by tarmac roads.

The snails were found in several colonies in each block. Allele frequencies for the gene *MDH-1* (alleles A and a) were obtained and compared. Statistical analysis of the allele frequencies of the two populations showed them to be significantly different ($P << 0.05$). Note: A Mann-Whitney *U* test was used in this instance. It is similar to a Student's *t* test, but does not assume a normal distribution of data (it is non-parametric).





	Colony	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15
Block A	*MDH-1* A %	39	39	36	42	39	47	32	42	44	42	44	50	50	58	75
	MDH-1 a %															
Block B	*MDH-1* A %	81	61	75	68	70	61	70	60	58	61	54	54	47		
	MDH-1 a %															

4. Complete the table above by filling in the frequencies of the *MDH-1* a allele:

5. Suggest why these snail populations are effectively geographically isolated: _____

_____

_____

_____

6. Both the *MDH-1* alleles produce fully operative enzymes. Suggest why the frequencies of the alleles have become significantly different.

_____

_____

_____

7. Identify the colony in block A that appears to be isolated from the rest of the block itself: _____

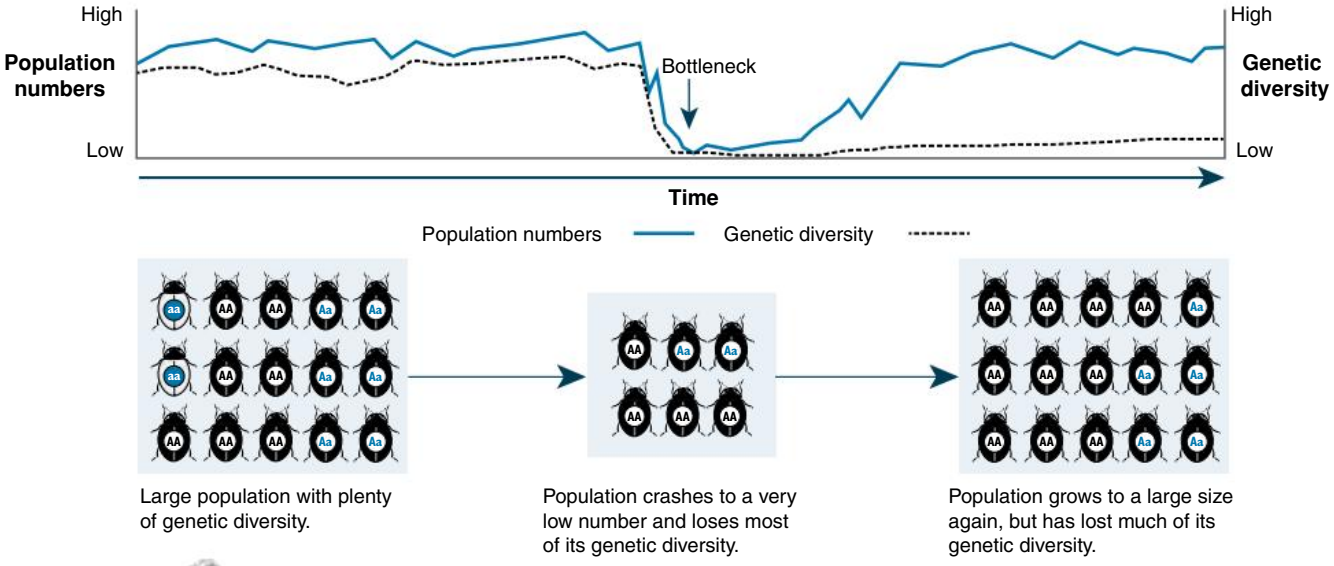
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220 Genetic Bottlenecks

Key Idea: Genetic bottlenecks occur when population numbers and diversity fall dramatically. Although a population's numbers may recover, its genetic diversity often does not.

Populations may sometimes be reduced to low numbers by predation, disease, or periods of climatic change. These large scale reductions are called genetic (or population) bottlenecks. The sudden population decline is not necessarily selective and it may affect all phenotypes equally. Large scale catastrophic events, such as fire or volcanic eruptions, are examples of such non-selective events. Affected populations may later recover, having squeezed through a 'bottleneck' of low numbers. The diagram below illustrates how population numbers may be reduced as a result of a catastrophic event. Following such an event, the gene pool of the surviving remnant population may be markedly different to that of the original gene pool. Genetic drift may cause further changes to allele frequencies. The small population may return to previous levels but with a reduced genetic diversity.

Change in population numbers and diversity





Modern Examples of Genetic Bottlenecks

Cheetahs: The world population of cheetahs currently stands at fewer than 20,000. Recent genetic analysis has found that the entire population exhibits very little genetic diversity. It appears that cheetahs may have narrowly escaped extinction at the end of the last ice age, about 10-20,000 years ago. If all modern cheetahs arose from a very limited genetic stock, this would explain their present lack of genetic diversity. The lack of genetic variation has resulted in a number of problems that threaten cheetah survival, including sperm abnormalities, decreased fecundity, high cub mortality, and sensitivity to disease.

Illinois prairie chicken: When Europeans first arrived in North America, there were millions of prairie chickens. As a result of hunting and habitat loss, the Illinois population of prairie chickens fell from about 100 million in 1900 to fewer than 50 in the 1990s. A comparison of the DNA from birds collected in the mid-twentieth century and DNA from the surviving population indicated that most of the genetic diversity has been lost.



Photo: Dept. of Natural Resources, Illinois

1. Endangered species are often subjected to genetic bottlenecks. Explain how genetic bottlenecks affect the ability of a population of an endangered species to recover from its plight:

2. Why has the lack of genetic diversity in cheetahs increased their sensitivity to disease?

3. Describe the effect of a genetic bottleneck on the potential of a species to adapt to changes (i.e. its ability to evolve):

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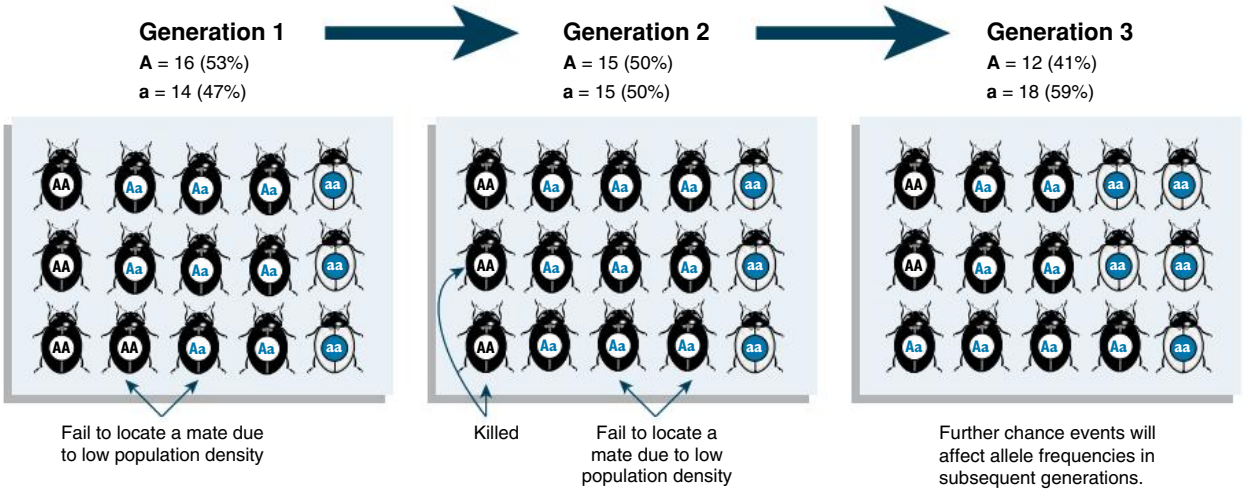
LINK 221 LINK 205 WEB 220 KNOW

221 Genetic Drift

Key Idea: Genetic drift is the term for the random changes in allele frequency that occur in all populations. It has a more pronounced effect in small populations.

Not all individuals, for various reasons, will be able to contribute their genes to the next generation. In a small population, the effect of a few individuals not contributing their alleles to the next generation can have a great effect on allele frequencies. Alleles may even become **lost** from the gene pool altogether (frequency becomes 0%) or **fixed** as the only allele for the gene present (frequency becomes 100%). The random change in allele frequencies is called **genetic drift**.

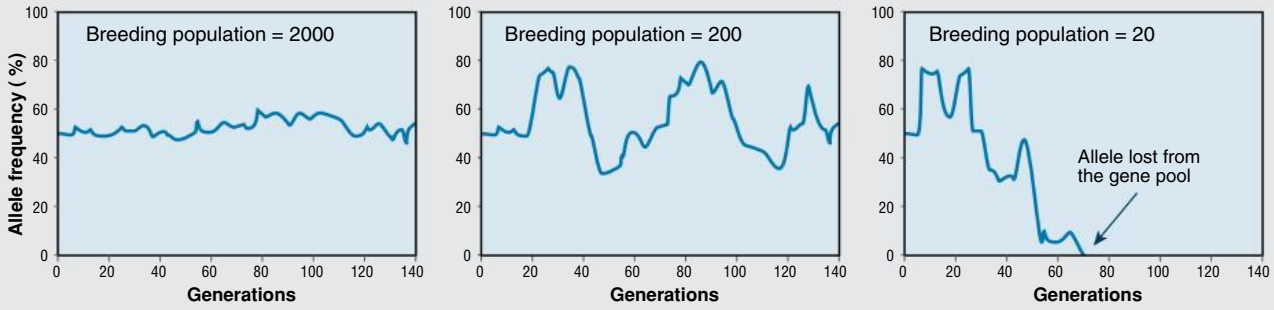
The genetic makeup (allele frequencies) of the population changes randomly over a period of time



This diagram shows the gene pool of a hypothetical small population over three generations. For various reasons, not all individuals contribute alleles to the next generation. With the random loss of the alleles carried by these individuals, the allele frequency changes from one generation to the next. The change in frequency is directionless as there is no selecting force. The allele combinations for each successive generation are determined by how many alleles of each type are passed on from the preceding one.

Computer Simulation of Genetic Drift

Below are displayed the change in allele frequencies in a computer simulation showing random genetic drift. The breeding population progressively gets smaller from left to right. Each simulation was run for 140 generations.



Large breeding population
Fluctuations are minimal in large breeding populations because the large numbers buffer the population against random loss of alleles. On average, losses for each allele type will be similar in frequency and little change occurs.

Small breeding population
Fluctuations are more severe in smaller breeding populations because random changes in a few alleles cause a greater percentage change in allele frequencies.

Very small breeding population
Fluctuations in very small breeding populations are so extreme that the allele can become fixed (frequency of 100%) or lost from the gene pool altogether (frequency of 0%).

1. (a) What is genetic drift? _____

_____

(b) Why is the effect of genetic drift more pronounced in small populations? _____

_____

2. Suggest why genetic drift is an important process in the evolution of small populations: _____

_____

_____

KNOW WEB 221 LINK 205 LINK 219

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222 KEY TERMS: Did You Get It?

1. Match each term to its definition, as identified by its preceding letter code.

adaptation .....

fitness .....

founder effect .....

gene pool .....

genetic bottleneck .....

mutation .....

new synthesis .....

phenotype .....

population .....

species .....

A The loss of genetic variation when a new colony is formed by a very small number of individuals from a larger population.

B An evolutionary event in which a significant proportion of the alleles in a population are lost.

C Observable characteristics in an organism.

D The union of the Darwin's ideas of evolution and Mendelian genetics, along with ideas from various branches of biology, to form a logical explanation of evolution.

E Group of organisms capable of breeding together to produce viable and fertile offspring.

F A measure of the capability of an organism with a certain genotype to reproduce. Usually expressed as a figure denoting the contribution of genes to the next generation.

G The total number of individuals of a species within a set habitat or area.

H A change to the DNA sequence of an organism. This may be a deletion, insertion, duplication, inversion or translocation of DNA in a gene or chromosome.

I The sum total of all genes of all breeding individuals in a population at any one time.

J Heritable characteristic of a species (behavioural, structural, or physiological) that equips it for survival and reproductive success in its environment.

2. Explain the characteristics of each of the following types of natural selection and state when each might operate:

(a) Directional selection: _____

_____

_____

(b) Stabilising selection: _____

_____

_____

(c) Disruptive selection: _____

_____

_____

3. Compare and contrast population bottlenecks with the founder effect. Explain why genetic drift is likely to be a more important phenomenon in small populations than larger ones. You may use extra paper if required.

_____

_____

_____

_____

_____

_____

_____

_____

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QLD As required
VCE Unit 4 (2)
WA ATAR Unit 3
SA Theme M, C

The Evidence for Evolution

Key terms

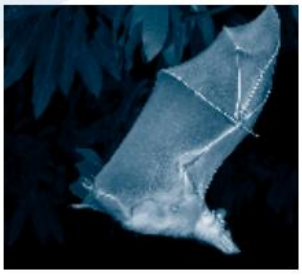
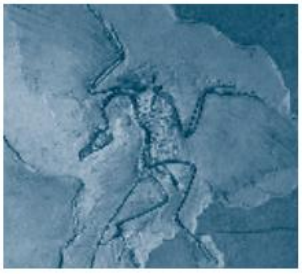
biogeographical evidence
chronometric dating (= absolute dating)
common ancestor
comparative anatomy
continental drift
evo-devo
fossil
fossil record
geologic time scale
homologous structure
Hox genes
molecular clock
palaeontological evidence
phylogeny
radiometric dating
relative dating
rock strata
transitional fossil

Comparative studies

Learning aims and skills

Activity number

- ☐ 1 Explain the biochemical evidence by the universality of DNA, amino acids, and protein structures (e.g. cytochrome c) for the common ancestry of living organisms. Describe how comparisons of specific molecules between species are used as an indication of relatedness or phylogeny. — 223-225
- ☐ 2 Discuss how biochemical variations can be used as a molecular clock to determine probable dates of divergence from a common ancestor. — 225
- ☐ 3 Explain how comparative anatomy and physiology have contributed to our understanding of evolutionary relationships. — 226 228
- ☐ 4 Explain how evolutionary developmental biology (evo-devo) has provided some of the strongest evidence for the mechanisms of evolution, particularly for the evolution of novel forms. — 164 227


Biogeographical evidence for evolution

Learning aims and skills

Activity number

- ☐ 5 Explain how founder populations may establish and evolve in isolation from their parent population. Describe features of typical oceanic island colonisers and give examples of where this has been important in the evolution of new species. — 229
- ☐ 6 Explain how the geographical distribution of living and extinct organisms provides evidence of dispersal from a point of origin across pre-existing barriers. — 231

The fossil record

Learning aims and skills

Activity number

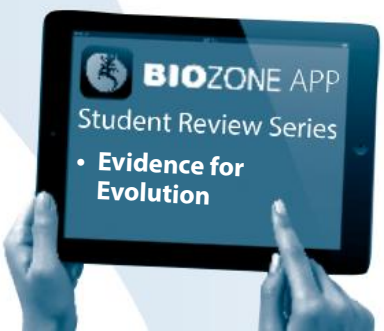
- ☐ 7 Outline the palaeontological evidence for evolution using an example, e.g. the evolution of horses or birds. — 228 230
- ☐ 8 Explain how fossils are formed and dated. Explain how the dating of fossil-bearing rocks has provided the data for dividing the history of life on Earth into geological periods, which collectively form the geologic time scale. — 230
- ☐ 9 Explain the significance of transitional fossils. Using examples, describe the trends that fossils indicate in the evolution of related groups. — 230

Modern examples of evolution

Learning aims and skills

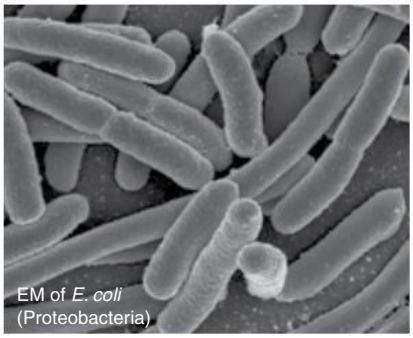
Activity number

- ☐ 10 Appreciate that evidence for evolution is not confined to the past. There are many documented examples of evolution occurring in the present day. Modern examples include the evolution of drug resistance in pathogens, the evolution of pesticide resistance in insects and many others. — 211-213 216-218



223 Descent and Common Ancestry

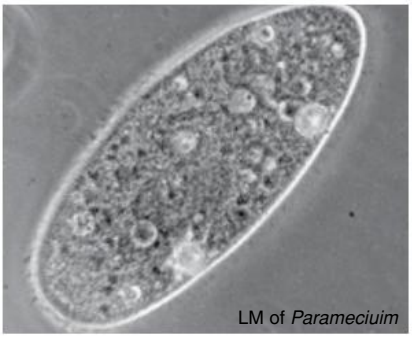
Key Idea: Key features shared by all life forms support the hypothesis that life on Earth evolved from a common ancestor. Our knowledge of how organisms are related has grown rapidly in recent decades thanks to new techniques in **molecular phylogenetics**. Such techniques compare the DNA, RNA, and proteins of organisms to establish evolutionary relationships. Molecular phylogenetics has enabled scientists to clarify the very earliest origins of eukaryotes and to recognise two prokaryote domains. Powerful evidence for the common ancestry of all life comes from the commonality in the genetic code, and from the similarities in the molecular machinery of all cells.



Rocky Mountain Laboratories, NIAID, NIH



Xiangyux (PD)



Barfooz and Josh Grosse (CC 3.0)

DOMAIN BACTERIA

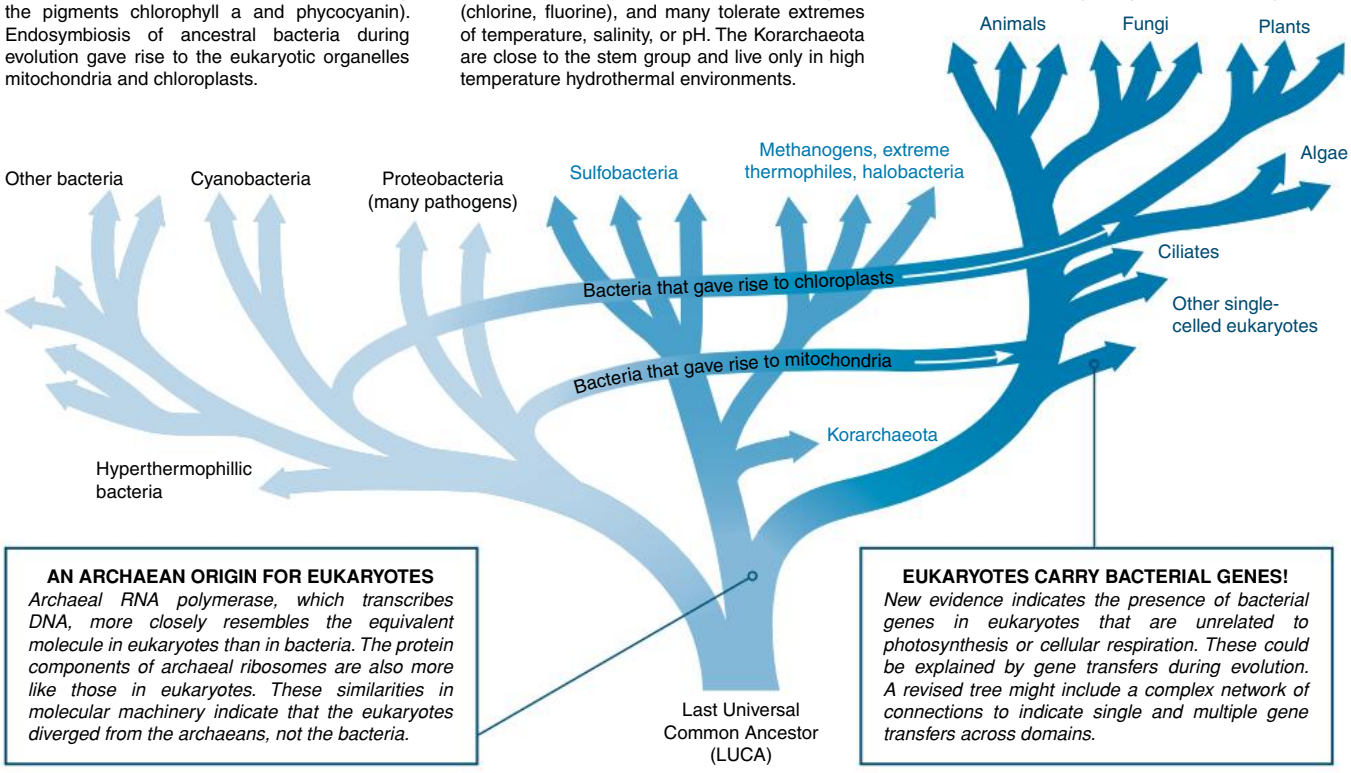
Lack a distinct nucleus and cell organelles. Generally prefer less extreme environments than Archaea. Includes well-known pathogens, many harmless and beneficial species, and the cyanobacteria (photosynthetic bacteria containing the pigments chlorophyll a and phycocyanin). Endosymbiosis of ancestral bacteria during evolution gave rise to the eukaryotic organelles mitochondria and chloroplasts.

DOMAIN ARCHAEA

Closely resemble eubacteria in many ways but membrane and cell wall composition and aspects of metabolism are very different. Live in extreme environments similar to those on primeval Earth. They may utilise sulfur, methane, or halogens (chlorine, fluorine), and many tolerate extremes of temperature, salinity, or pH. The Korarchaeota are close to the stem group and live only in high temperature hydrothermal environments.

DOMAIN EUKARYA

Complex cells with organelles and a membrane-bound nucleus. This domain contains four of the kingdoms recognised under a traditional scheme. Note that Kingdom Protista is separated into distinct taxa, recognising their diverse origins.



AN ARCHAEAN ORIGIN FOR EUKARYOTES

Archaeal RNA polymerase, which transcribes DNA, more closely resembles the equivalent molecule in eukaryotes than in bacteria. The protein components of archaeal ribosomes are also more like those in eukaryotes. These similarities in molecular machinery indicate that the eukaryotes diverged from the archaeans, not the bacteria.

EUKARYOTES CARRY BACTERIAL GENES!

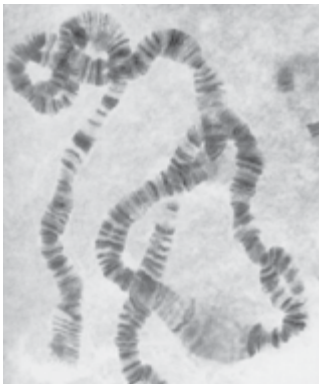
New evidence indicates the presence of bacterial genes in eukaryotes that are unrelated to photosynthesis or cellular respiration. These could be explained by gene transfers during evolution. A revised tree might include a complex network of connections to indicate single and multiple gene transfers across domains.

Adapted from: Uprooting the tree of life (see Appendix)

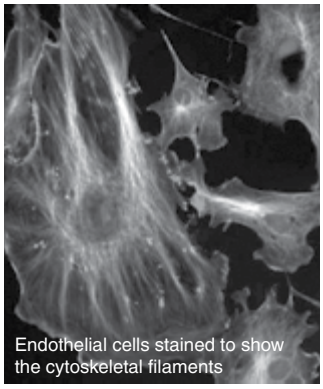
1. Explain the role of molecular phylogenetics in revising the traditional classification schemes (pre-1980):

2. Describe the evidence for the archaean origin of eukaryotic cells:

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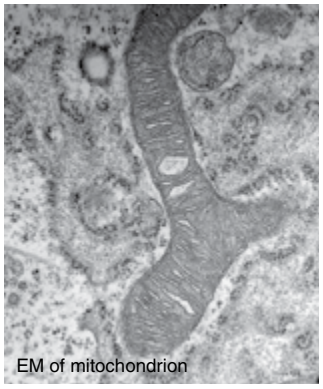

RGN

Eukaryotic cells are characterised by large linear chromosomes within the cell's nucleus (above). These nuclear chromosomes are packaged by proteins into a condensed structure called chromatin. The evolution of linear chromosomes was related to the appearance of microtubule-dependent cell division (mitosis and meiosis).



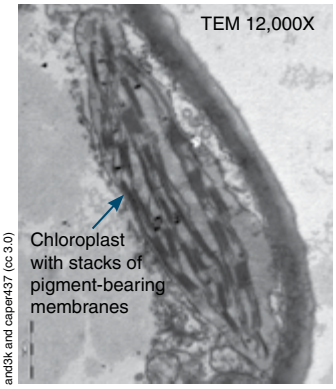
NIH (PD)

The cytoskeleton (CSK) is an intracellular scaffold of protein filaments, important in intracellular movement of materials and cell division in eukaryotic cells. The CSK was once thought to be unique to eukaryotic cells, but homologues to all the major proteins of the eukaryotic cytoskeleton have also been found in prokaryotes.



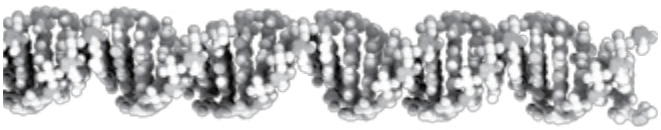
EII

The endosymbiotic origin of eukaryotic mitochondria is supported by the evidence from mitochondrial gene sequences, ribosomes, and protein synthesis that indicate a prokaryotic origin. Mitochondria are characteristic of eukaryotic cells and arose in a common ancestor to eukaryotic cells.



and3k and caper437 (cc 3.0)

Chloroplasts are assumed to have been acquired, via endosymbiosis, after mitochondria and there is much evidence to suggest that they were acquired independently by more than one type of cyanobacterial cell (i.e. their origin is polyphyletic). The prokaryotic origin of chloroplasts is supported by ribosomal evidence.



Most organisms share the same genetic code, i.e. the same combination of three DNA bases code for the same amino acid, although there are some minor variations (e.g. in mitochondria). Evidence suggests the code was subject to selection pressure which acted to minimise the effect of point mutations or errors in translation.



EII

In all living systems, the genetic machinery consists of self-replicating DNA molecules. Some DNA is transcribed into RNA, some of which is translated into proteins. The machinery for translation (above) involves proteins and RNA. Ribosomal RNA analyses support a universal common ancestor.

3. (a) Describe how eukaryotic organelles, such as mitochondria and chloroplasts, are thought to have evolved:

(b) What evidence is there to support this?

4. Discuss the evidence for a Last Universal Common Ancestor?

5. Explain how molecular phylogenetics has helped us to understand the diversity on Earth and the relatedness of all life:

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224 Homologous DNA Sequences

Key Idea: The relatedness of species can be deduced from the differences in their DNA.

DNA hybridisation (below) provides a way to compare the genomes of different species by measuring the degree of genetic similarity between DNA sequences. More closely related species have fewer differences between their genomes than more distantly related species. This technique gives a measure of 'relatedness', and can be calibrated as a **molecular clock** against known fossil dates. It has been used to help determine the approximate date of human divergence from the apes, which has been estimated to be between 10 and 5 million years ago.

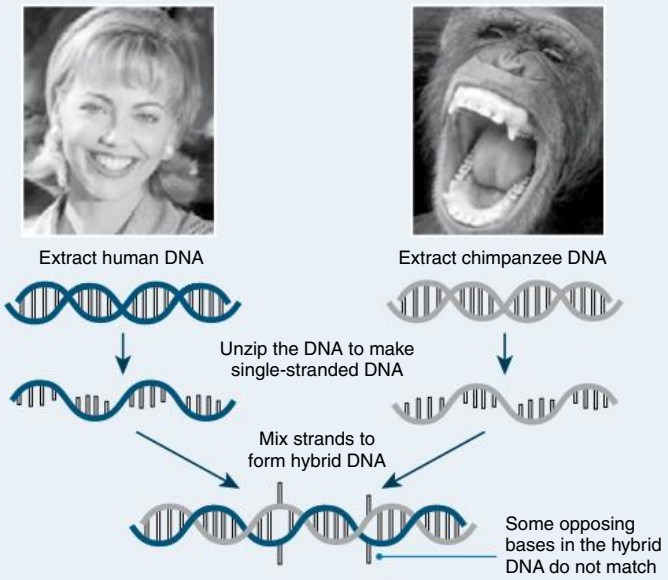
DNA Hybridisation

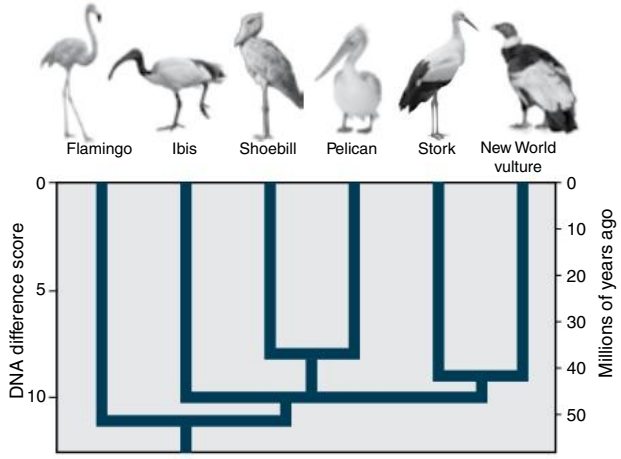
1. DNA from the two species to be compared is extracted, purified and cut into short fragments (e.g. 600-800 base pairs).
2. The DNA of one species is mixed with the DNA of another.
3. The mixture is incubated to allow DNA strands to dissociate and reanneal, forming hybrid double-stranded DNA.
4. The hybridised sequences that are highly similar will bind more firmly. A measure of the heat energy required to separate the hybrid strands provides a measure of DNA relatedness.

DNA Homologies Today

DNA-DNA hybridisation has been criticised because duplicated sequences within a single genome make it unreliable comparing closely related species.

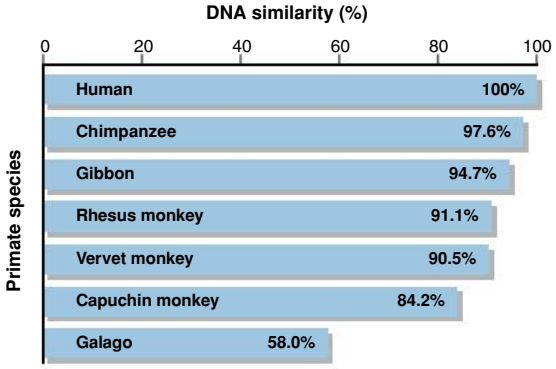
Today, DNA sequencing and computed comparisons are more widely used to compare genomes, although DNA-DNA hybridisation is still used to help identify bacteria.





The relationships among the New World vultures and storks have been determined using DNA hybridisation. It has been possible to estimate how long ago various members of the group shared a common ancestor.

Similarity of human DNA to that of other primates



The genetic relationships among the primates has been investigated using DNA hybridisation. Human DNA was compared with that of the other primates. It largely confirmed what was suspected from anatomical evidence.

1. Explain how **DNA hybridisation** can give a measure of genetic relatedness between species:

_____

_____

_____

2. Study the graph showing the results of a DNA hybridisation between human DNA and that of other primates.

   (a) Which is the most closely related primate to humans? _____

   (b) Which is the most distantly related primate to humans? _____

3. State the DNA difference score for: (a) Shoebills and pelicans:_____ (b) Storks and flamingos: _____

4. On the basis of DNA hybridisation, state how long ago the ibises and New World vultures shared a common ancestor:

_____

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225 Homologous Proteins

Key Idea: Comparative biochemistry provides new tools for identifying relationships between species.

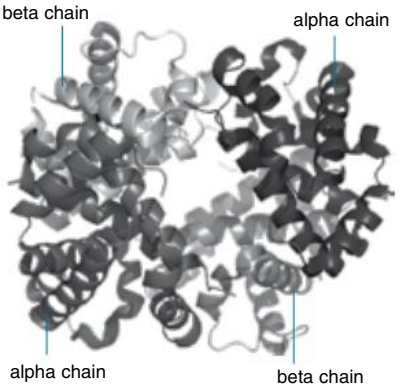
Traditionally, phylogenies were based largely on anatomical or behavioural traits and biologists attempted to determine the relationships between organisms based on overall degree of similarity or by tracing the appearance of key characteristics. With the advent of molecular techniques, **homologies** (similarities resulting from shared ancestry) can now be studied at the molecular level as well, and these can be compared to the phylogenies established using other methods. Protein sequencing provides an excellent tool for establishing homologies. A protein has a specific number of amino acids arranged in a specific order. Any differences in the sequence reflect changes in the DNA sequence. Commonly studied proteins include blood proteins, such as haemoglobin, and the respiratory protein cytochrome *c*.

Amino Acid Differences in Haemoglobin

Human beta chain	0
Chimpanzee	0
Gorilla	1
Gibbon	2
Rhesus monkey	8
Squirrel monkey	9
Dog	15
Horse, cow	25
Mouse	27
Grey kangaroo	38
Chicken	45
Frog	67

When the sequence of the **beta haemoglobin chain** (right), which is 146 amino acids long, is compared between humans, five other primates, and six other vertebrates, the results support the phylogenies established using other methods. The numbers in the table (left) represent the number of amino acid differences between the beta chain of humans and those of other species. In general, the number of amino acid differences between the haemoglobins of different vertebrates is inversely proportional to genetic relatedness.

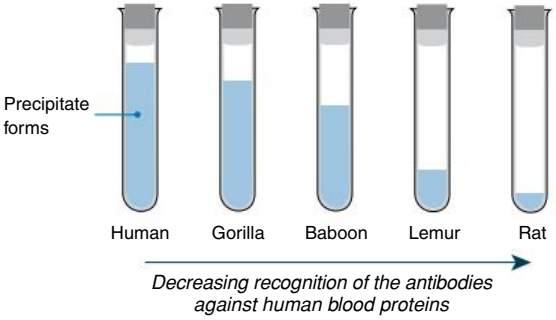
Shading indicates (from top) primates, non-primate placental mammals, marsupials, and non-mammals.

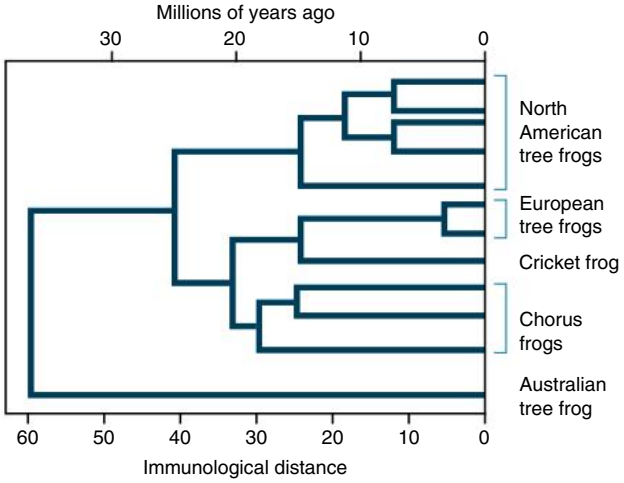


In most vertebrates, the oxygen-transporting blood protein haemoglobin is composed of four polypeptide chains, two alpha chains and two beta chains. Haemoglobin is derived from myoglobin, and ancestral species had just myoglobin for oxygen transport. When the amino acid sequences of myoglobin, the haemoglobin alpha chain, and the haemoglobin beta chain are compared, there are several amino acids that remain conserved between all three. These amino acid sequences must be essential for function because they have remained unchanged throughout evolution.

Using Immunology to Determine Phylogeny

The immune system of one species will recognise the blood proteins of another species as foreign and form antibodies against them. This property can be used to determine the extent of relatedness between species. Blood proteins, such as albumins, are used to prepare **antiserum** in rabbits. The antiserum contains antibodies against the test blood proteins (e.g. human) and will react to those proteins in any blood sample they are mixed with. The extent of the reaction indicates how similar the proteins are; the greater the reaction, the more similar the proteins. This principle is illustrated (right) for antiserum produced to human blood and its reaction with the blood of other primates and a rat.







The relationships among tree frogs have been established by immunological studies based on blood proteins such as immunoglobulins and albumins. The **immunological distance** is a measure of the number of amino acid substitutions between two groups. This, in turn, has been calibrated to provide a time scale showing when the various related groups diverged.

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Cytochrome *c* and the Molecular Clock Theory

Evolutionary change at the molecular level occurs primarily through fixation of neutral mutations by genetic drift. The rate at which one neutral mutation replaces another depends on the mutation rate, which is fairly constant for any particular gene.

If the rate at which a protein evolves is roughly constant over time, the amount of molecular change that a protein shows can be used as a molecular clock to date evolutionary events, such as the divergence of species.

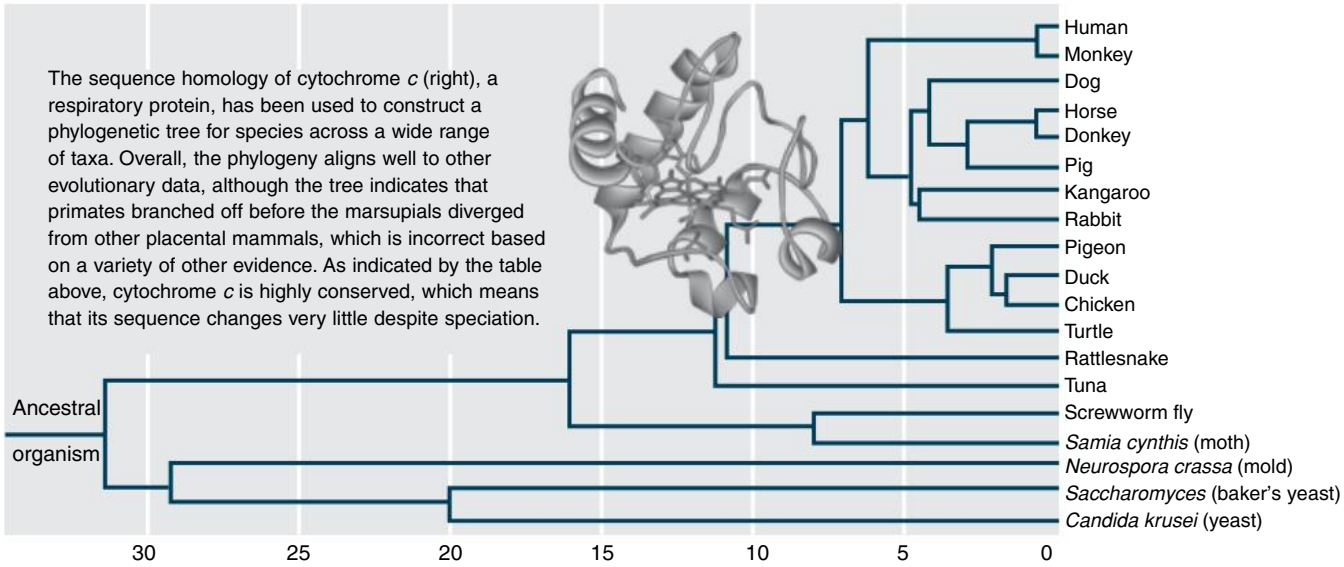
The molecular clock for each species, and each protein, may run at different rates, so scientists calibrate the molecular clock data with other evidence (morphological, molecular) to confirm phylogenetic relationships.

For example, 20 amino acid substitutions in a protein since two organisms diverged from a known common ancestor 400 mya indicates an average substitution rate of 5 substitutions per 100 my.

		1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22
Human		Gly	Asp	Val	Glu	Lys	Gly	Lys	Lys	Ile	Phe	Ile	Met	Lys	Cys	Ser	Gln	Cys	His	Thr	Val	Glu	Lys
Pig												Val	Gln			Ala							
Chicken				Ile						Val		Val	Gln			Ala							
Dogfish										Val		Val	Gln			Ala							Asn
Drosophila	<<									Leu		Val	Gln	Arg		Ala							Ala
Wheat	<<		Asn	Pro	Asp	Ala		Ala				Lys	Thr	Arg		Ala						Asp	Ala
Yeast	<<		Ser	Ala	Lys			Ala	Thr	Leu		Lys	Thr	Arg		Glu	Leu						

This table shows the N-terminal 22 amino acid residues of human cytochrome *c*, with corresponding sequences from other organisms aligned beneath. Sequences are aligned to give the most position matches. A shaded square indicates no change. In every case, the cytochrome's heme group is attached to the Cys-14 and Cys-17. In *Drosophila*, wheat, and yeast, arrows indicate that several amino acids precede the sequence shown.

The sequence homology of cytochrome *c* (right), a respiratory protein, has been used to construct a phylogenetic tree for species across a wide range of taxa. Overall, the phylogeny aligns well to other evolutionary data, although the tree indicates that primates branched off before the marsupials diverged from other placental mammals, which is incorrect based on a variety of other evidence. As indicated by the table above, cytochrome *c* is highly conserved, which means that its sequence changes very little despite speciation.



1. Explain why chimpanzees and gorillas are considered most closely related to humans, while monkeys are less so:

2. (a) Why would a respiratory protein like cytochrome *c* be highly conserved?

   (b) Why are highly conserved proteins good candidates for use in establishing phylogenies?

3. Discuss some of the limitations of using protein homology, specifically molecular clocks, to establish phylogeny:

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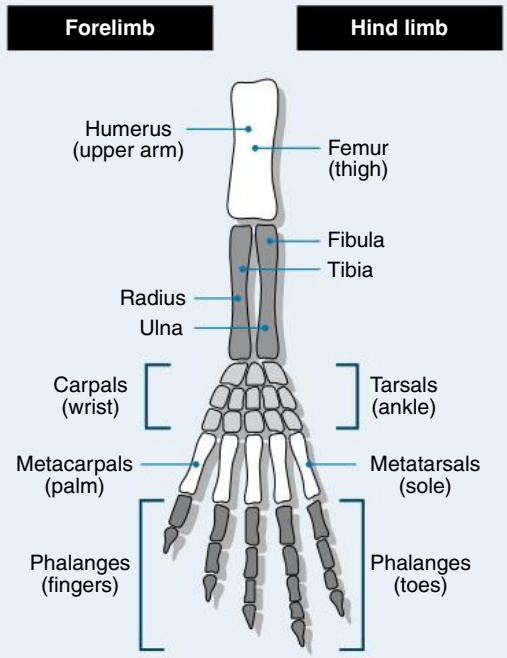
226 Homologous Structures

Key Idea: Homologous structures (homologies) are structural similarities present as a result of common ancestry. The common structural components have been adapted to different purposes in different taxa.

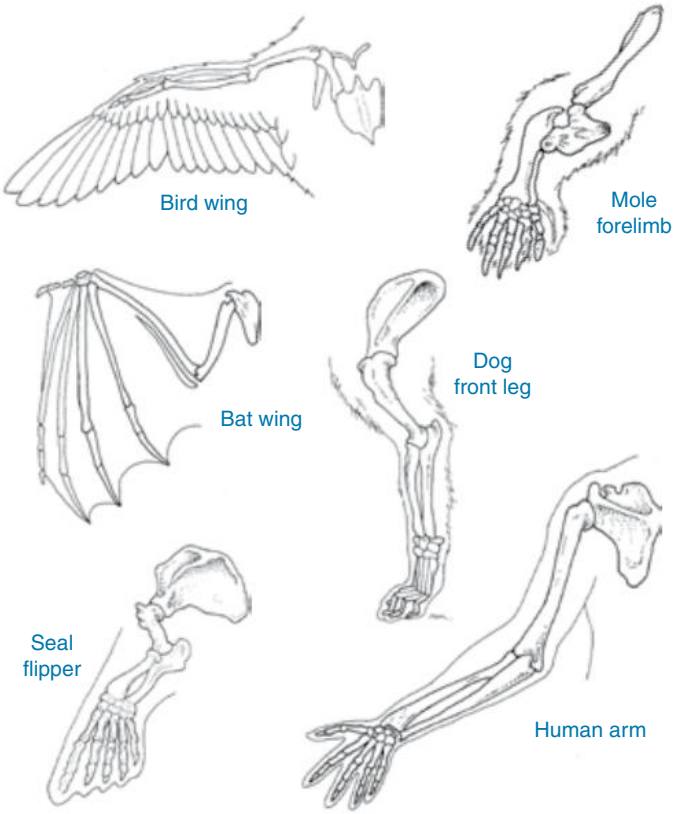
The bones of the forelimb of air-breathing vertebrates are composed of similar bones arranged in a comparable pattern. This is indicative of common ancestry. The early land vertebrates were amphibians with a **pentadactyl limb** structure (a limb with five fingers or toes). All vertebrates that descended from these early amphibians have limbs with this same basic pentadactyl pattern. They also illustrate the phenomenon known as **adaptive radiation**, since the basic limb plan has been adapted to meet the requirements of different niches.

Generalised Pentadactyl Limb

The forelimbs and hind limbs have the same arrangement of bones but they have different names. In many cases, the basic limb plan has been adapted (e.g. by loss or fusion of bones) to meet the requirements of different niches (e.g. during adaptive radiation of the mammals).



Specialisations of Pentadactyl Limbs



1. Briefly describe the purpose of the major anatomical change that has taken place in each of the limb examples above:

   (a) Bird wing: Highly modified for flight. Forelimb is shaped for aerodynamic lift and feather attachment.

   (b) Human arm: _____

   (c) Seal flipper: _____

   (d) Dog front leg: _____

   (e) Mole forelimb: _____

   (f) Bat wing: _____

2. Explain how homology in the pentadactyl limb is evidence for adaptive radiation: _____

3. Homology in the behaviour of animals (for example, sharing similar courtship or nesting rituals) is sometimes used to indicate the degree of relatedness between groups. How could behaviour be used in this way:

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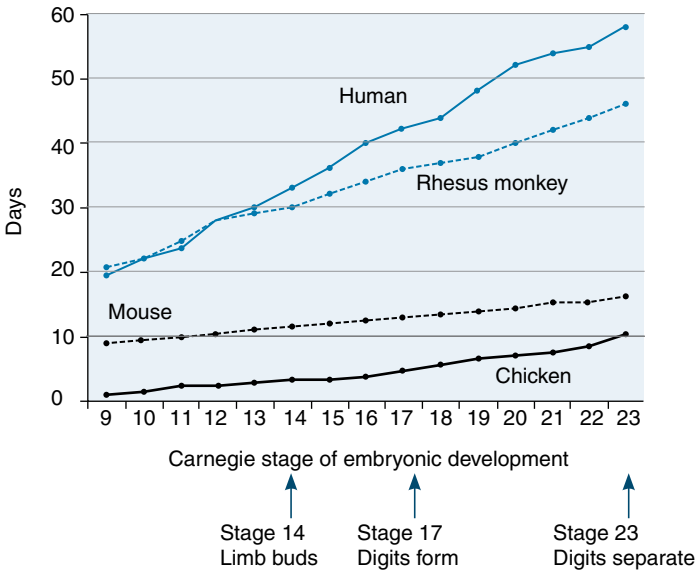
227 Developmental Evidence for Evolution

Key Idea: Similarities in the development of embryos, including the genetic control of that development, provides strong evidence for evolution.

Developmental biology studies the processes by which organisms grow and develop. In the past, it was restricted to the appearance of a growing embryo. Today, developmental biology focuses on the genetic control of development and its role in producing the large differences we see in the adult appearance of different species. Differences in gene expression during development are behind these differences.

Developmental biology

During development, vertebrate embryos pass through the same stages, in the same sequence, regardless of the total time period of development. This similarity is strong evidence of their shared ancestry. The stage of embryonic development is identified using a standardised system based on the development of structures, not by size or the number of days of development. The Carnegie stages (right) cover the first 60 days of development.



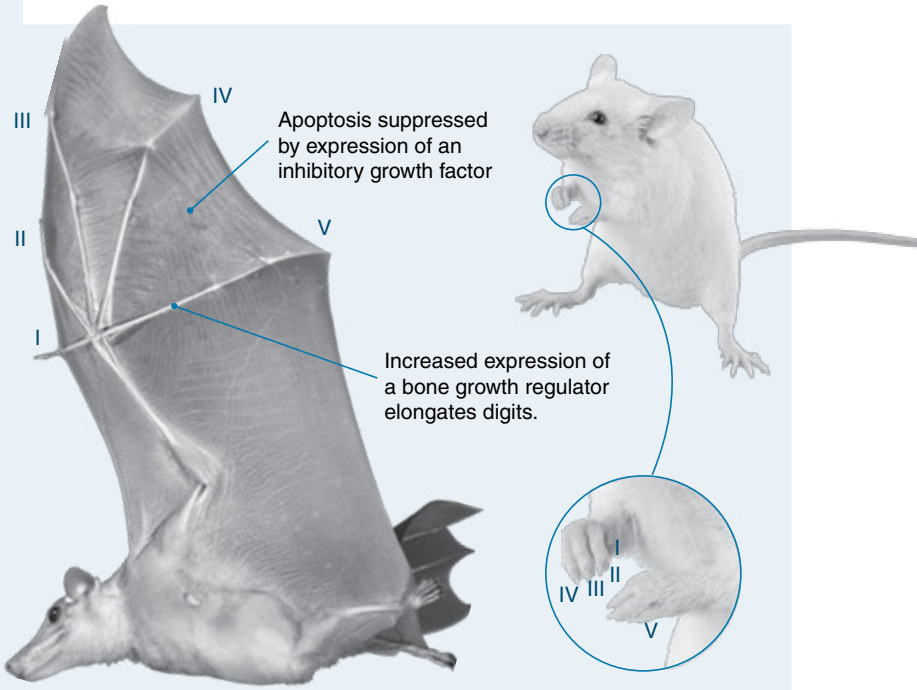
Limb homology and the control of development

As we have seen, homology (e.g. in limb structure) is evidence of shared ancestry. How do these homologous structures become so different in appearance? The answer lies in the way the same genes are regulated during development.

All vertebrate limbs form as buds at the same stage of development. At first, the limbs resemble paddles, but **apoptosis** (programmed cell death) of the tissue between the developing bones separates the digits to form fingers and toes.

Like humans, mice have digits that become fully separated by interdigital apoptosis during development. In bat forelimbs, this controlled destruction of the tissue between the forelimb digits is inhibited. The developmental program is the result of different patterns of expression of the same genes in the two types of embryos.

Bat wings are highly specialised structures with unique features, such as elongated wrist and fingers (I-V) and membranous wing surfaces. The forelimb structures of bats and mice are homologous, but how the limb looks and works is quite different.



1. Describe a feature of vertebrate embryonic development that supports evolution from a common ancestor:

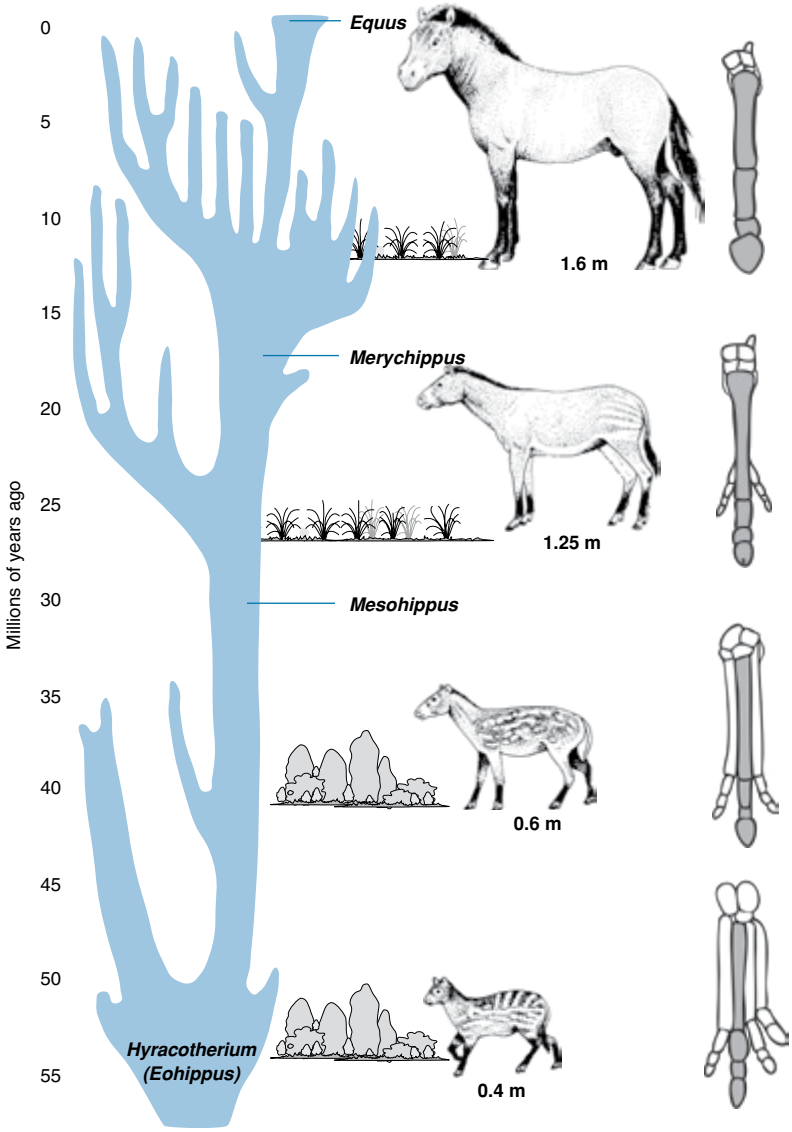
2. Explain how different specialised limb structures can arise from a basic pentadactyl structure:

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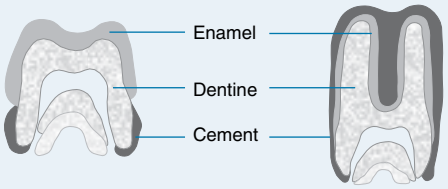
228 The Evolution of Horses

Key Idea: The evolution of the horse is one of the most robust examples of evolution documented in the fossil record.

The evolution of the horse from the ancestral *Hyracotherium* to modern *Equus* is well documented in the fossil record. The rich fossil record, which includes numerous **transitional fossils**, has enabled scientists to develop a robust model of horse phylogeny. It is a complex tree-like lineage with many divergences (below), and a diverse array of often coexisting species. The environmental transition from forest to grasslands drove many of the changes observed in the fossil record. These include reduction in toe number, increased size of cheek teeth, and increasing body size.



The cooler climates that prevailed in the Miocene (23 -5 mya) brought about a reduction in forested areas with grasslands becoming more abundant. The change in vegetation resulted in the equids developing more durable teeth to cope with the harsher diet. Over time the equid molar became longer and squarer with a hard cement-like covering to enable them to grind the grasses which became their primary diet.



Hyracotherium molar — *Equus* molar

The equids also became taller and faster to enable them to view and escape their predators. This is evident in their overall increase in size and the elongation of their limbs. The reduction in the number of toes from four to one (left) also enabled them to run faster and more efficiently.



The majority of equid evolution took place in North America, although now extinct species did migrate to other areas of the globe at various times. During the late Pliocene (2.6 mya) *Equus* spread into the Old World and diversified into several species including the modern zebra of Africa and the true horse, *Equus caballus*. Ironically, the horse became extinct in the Americas about 11,000 years ago, and was reintroduced in the 16th century by Spanish explorers.

1. Explain how the environmental change from forest to grassland influenced the following aspects of equid evolution:

   (a) Change in tooth structure: _____

   _____

   (b) Limb length: _____

   _____

   (c) Reduction in number of toes: _____

   _____

2. In which way does the equid fossil record provide a good example of the evolutionary process? _____

   _____

   _____

   _____

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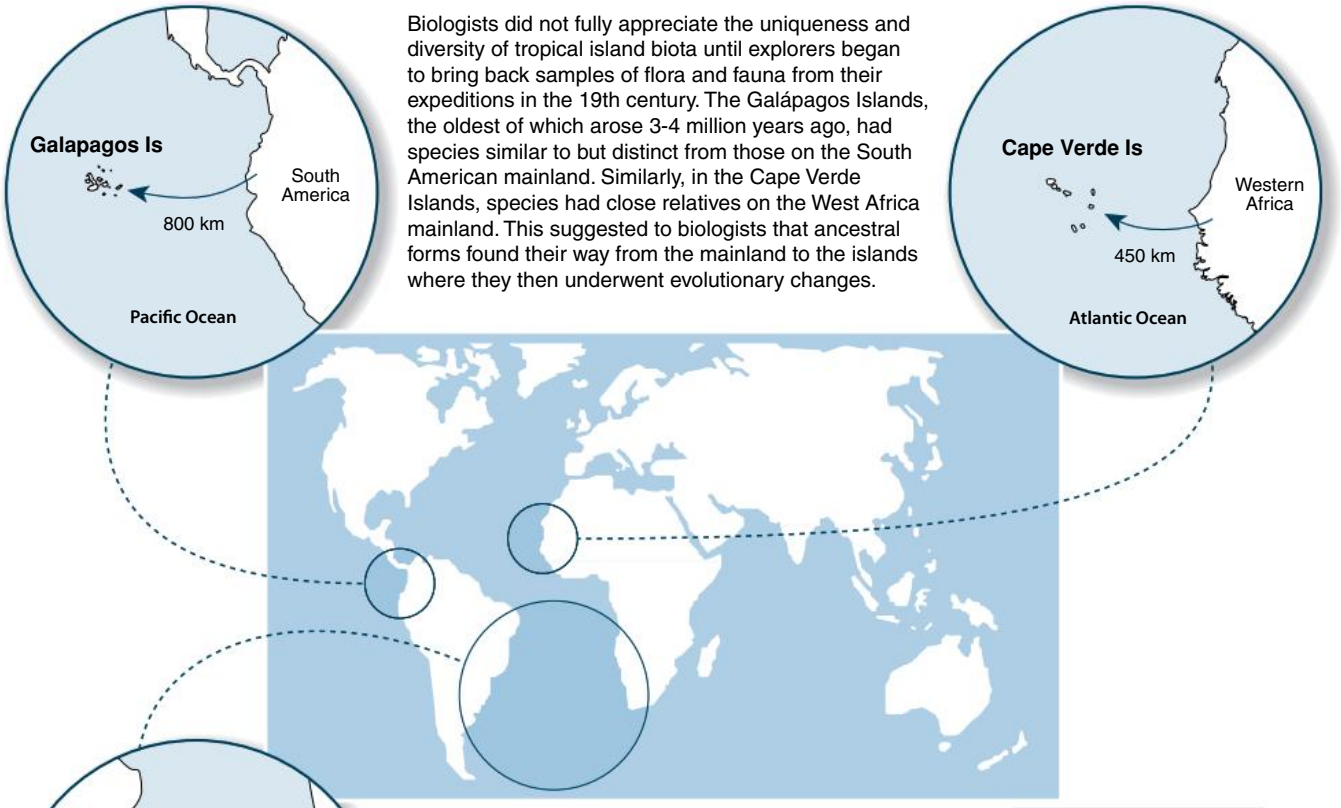
229 Oceanic Island Colonisers

Key Idea: Biogeography provides evidence for how the evolution of species is influenced by isolation, continental influences, geological processes, and dispersal ability.

Biogeography is the study of the geographical distribution of species. As described in this activity, island biogeography demonstrates that species on a given island may more closely resemble species on a nearby mainland, rather than species on a distant island with a similar environment.

Galápagos and Cape Verde islands

Biologists did not fully appreciate the uniqueness and diversity of tropical island biota until explorers began to bring back samples of flora and fauna from their expeditions in the 19th century. The Galápagos Islands, the oldest of which arose 3-4 million years ago, had species similar to but distinct from those on the South American mainland. Similarly, in the Cape Verde Islands, species had close relatives on the West Africa mainland. This suggested to biologists that ancestral forms found their way from the mainland to the islands where they then underwent evolutionary changes.



Tristan da Cunha

The island of Tristan da Cunha in the South Atlantic Ocean is a great distance from any other land mass. Even though it is closer to Africa, there are more species closely related to South American species found there (see table on right). This is probably due to the predominant westerly trade winds from the direction of South America. The flowering plants of universal origin are found in both Africa and South America and could have been introduced from either land mass.

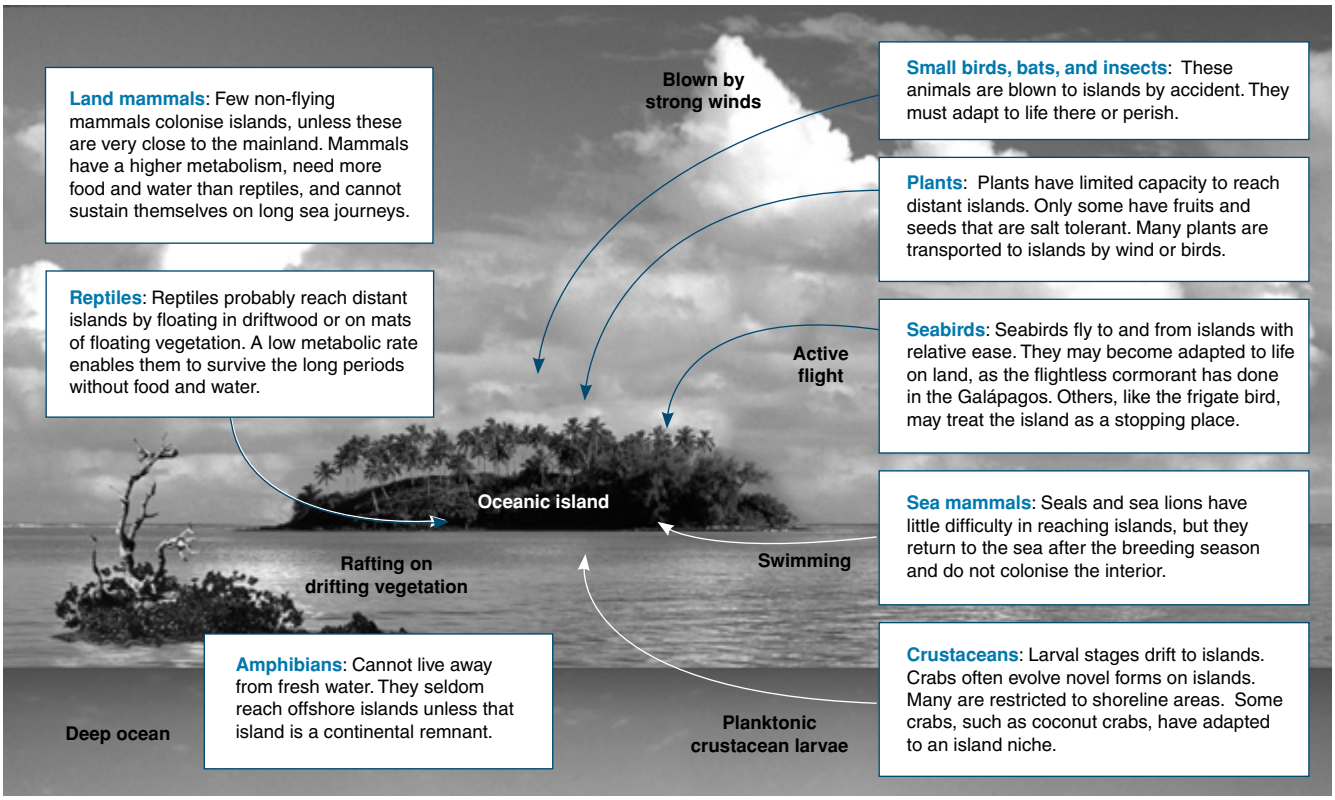
Tristan da Cunha species	
South American origin	
7	Flowering plants
5	Ferns
30	Liverworts
African origin	
2	Flowering plants
2	Ferns
5	Liverworts
Universal origin	
19	Flowering plants



The flightless cormorant (above) is one of a number of bird species that lost the power of flight after becoming an island resident. Giant tortoises, such as the 11 subspecies remaining on the Galápagos today (centre) were, until relatively recently, characteristic of many islands in the Indian Ocean including the Seychelles archipelago, Reunion, Mauritius, Farquhar, and Diego Rodriguez. These were almost completely exterminated by early Western sailors, although a small population remains on the island of Aldabra. Another feature of oceanic islands is the adaptive radiation of colonising species into different specialist forms. The three species of Galápagos iguana almost certainly arose, through speciation, from a hardy traveller from the South American mainland. The marine iguana (above) feeds on shoreline seaweeds and is an adept swimmer. The two species of land iguana (not pictured) feed on cacti, which are numerous. One of these (the pink iguana) was identified as a separate species only in 2009.

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The diversity and uniqueness of island biota is determined by migration to and from the island and extinctions and diversifications following colonisation. These events are themselves affected by a number of other factors (see table, below right). The organisms that successfully colonise islands have to be marine in habit, or able to survive long periods at sea or in the air. The biota of the **Galápagos Islands** provide a good example of the results of such a colonisation process. For example, all the subspecies of giant tortoise evolved in Galápagos from a common ancestor that arrived from the mainland, floating with the ocean currents.

Factors affecting final biota
Degree of isolation
Length of time of isolation
Size of island
Climate (tropical/Arctic, arid/humid)
Location relative to ocean currents
Initial plant and animal composition
The species composition of earliest arrivals (if always isolated)
Serendipity (chance arrivals)

1. The Galápagos and the Cape Verde Islands are both tropical islands close to the equator, yet their biotas are quite different. Explain why this is the case:

_____

_____

_____

2. Explain why the majority of the plant species found on Tristan da Cunha originated from South America, despite its greater distance from the island:

_____

_____

3. The table (right) identifies some of the factors influencing the composition of island biota. Explain how each of the three following might affect the diversity and uniqueness of the biota found on an oceanic island:

(a) Large island area: _____

_____

(b) Long period of isolation from other land masses: _____

_____

(c) Relatively close to a continental land mass: _____

_____

4. Describe one feature typical of an oceanic island coloniser and explain its significance: _____

_____

_____

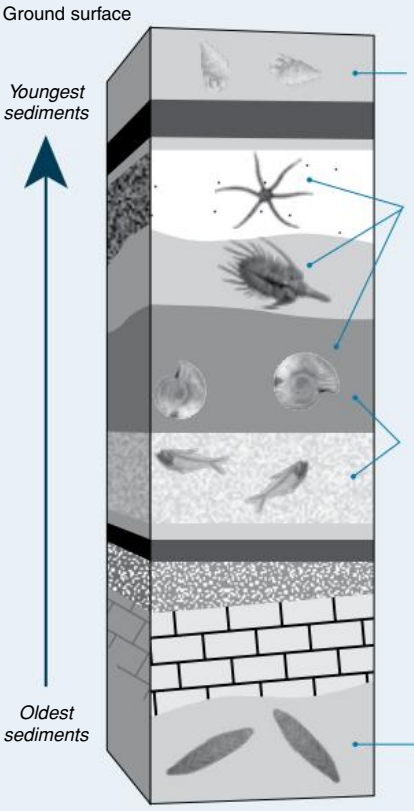
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230 Relative Dating and the Fossil Record

Key Idea: Fossils found in rock at the bottom of a rock profile are older than those found at the top of the rock file.

Relative dating establishes the sequential (relative) order of past events in a rock profile, but it can not provide an absolute date for an event. Each rock layer (**stratum**) is unique in terms of the type of rock (sedimentary or volcanic) and the type of fossils it contains. Rock layers (**strata**) are arranged in the order that they were deposited, with the oldest layers at the bottom (unless they have been disturbed by geological events). Strata from widespread locations can be correlated because a particular stratum at one location is the same age as the same stratum at a different location.

Profile with Sedimentary Rocks Containing Fossils



Recent fossils are found in more recent sediments
The more recent the layer of rock, the more resemblance there is between the fossils found in it and living forms.

Numerous extinct species
The number of extinct species is enormously greater than the number living today.

Fossil types differ in each sedimentary rock layer
Fossils found in a given layer of sedimentary rock generally differ in significant respects from those in other layers.

Only primitive fossils are found in older sediments
Phyla are represented by more generalised forms in the older layers, and not by specialised forms (such as those alive today).

New fossil types mark changes in environment

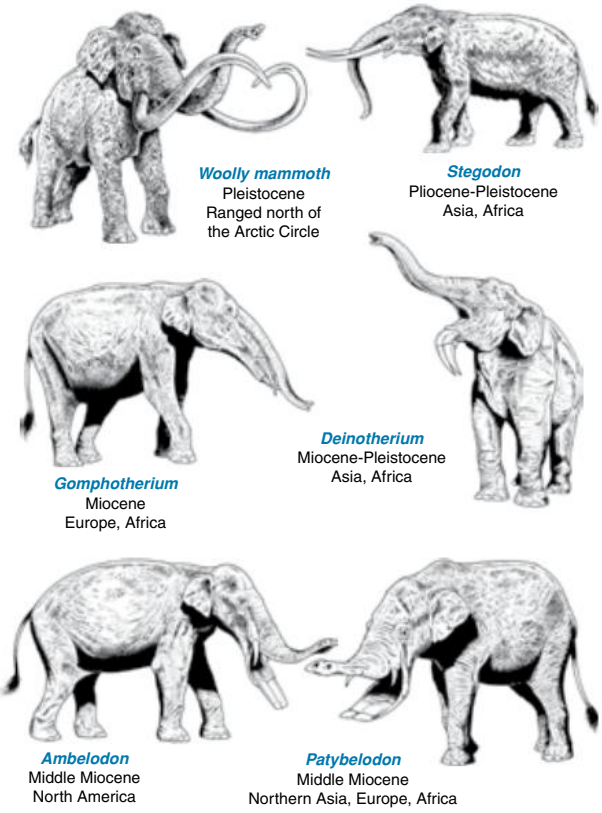
In the rocks marking the end of one geologic period, it is common to find many new fossils that become dominant in the next. Each geologic period had an environment very different from those before and after. Their boundaries coincided with drastic environmental changes and the appearance of new niches. These produced new selection pressures, resulting in new adaptive features in the surviving species as they responded to the changes. An absolute (chronometric) age can be assigned to fossils, usually by dating the rocks around them. Most often, these chronometric dating methods involve radiometric dating (e.g. radiocarbon, K-Ar).

The Rate of Evolution

According to the fossil record, rates of evolutionary change seem to vary. There are bursts of species formation and long periods of relative stability within species (stasis). The occasional rapid evolution of new forms apparent in the fossil record is probably a response to a changing environment. During periods of stable environmental conditions, the rate of evolutionary change may slow.

The Fossil Record of Proboscidea

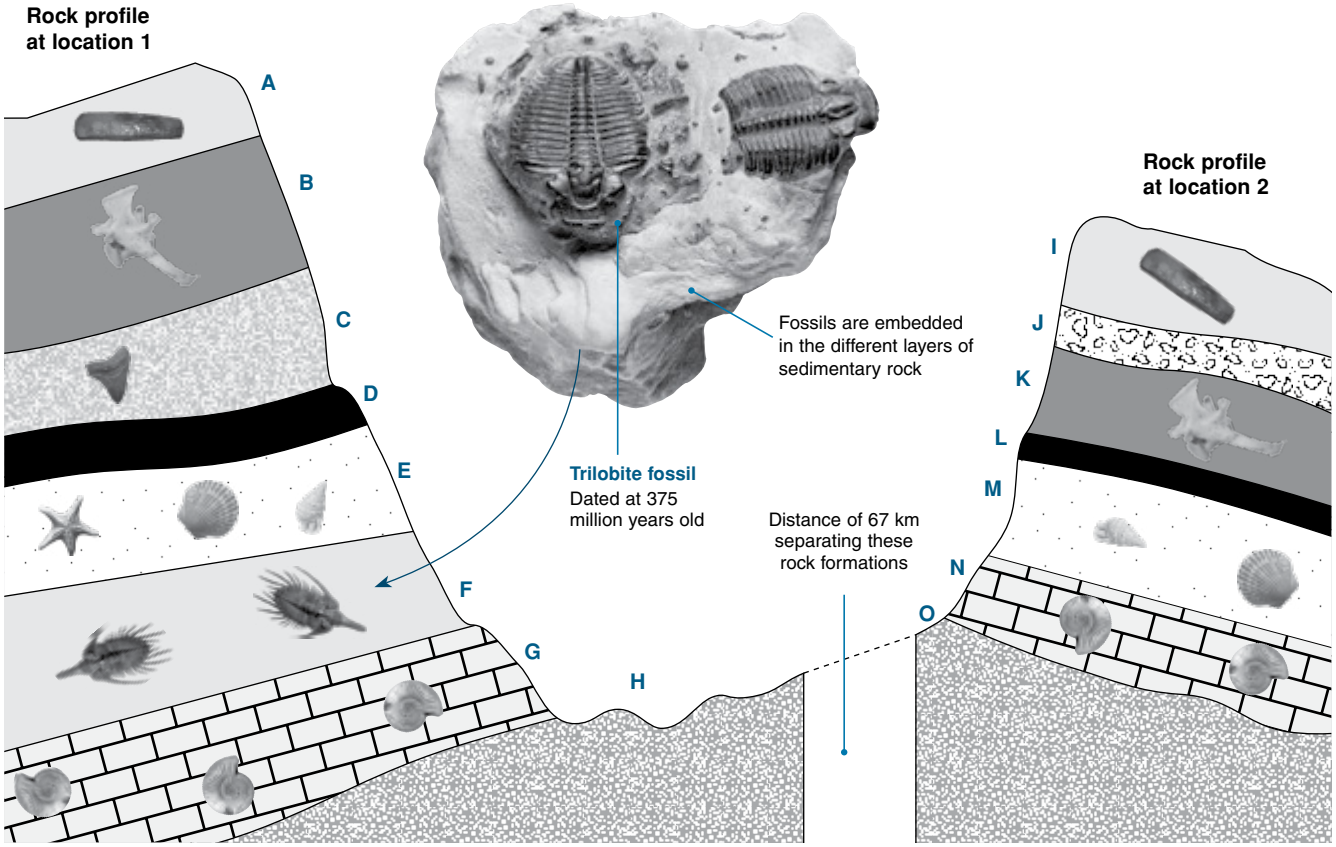
African and Indian elephants have descended from a diverse group known as **proboscideans** (named for their long trunks). The first pig-sized, trunkless members of this group lived in Africa 40 million years ago. From Africa, their descendants invaded all continents except Antarctica and Australia. As the group evolved, they became larger, an effective evolutionary response to deter predators. Examples of extinct members of this group are illustrated below:



- **Modern day species can be traced:** The evolution of many present-day species can be very well reconstructed. For instance, the evolutionary history of the modern elephants is exceedingly well documented for the last 40 million years. The modern horse also has a well understood fossil record spanning the last 50 million years.
- **Fossil species are similar to but differ from today's species**: Most fossil animals and plants belong to the same major taxonomic groups as organisms living today. However, they do differ from the living species in many features.

1. Name an animal or plant taxon (e.g. family, genus, or species) that has:
   (a) A good fossil record of evolutionary development: _____
   (b) Appeared to have changed very little over the last 100 million years or so: _____
2. Explain the importance of **fossils** as a record of evolutionary change over time: _____

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The questions below relate to the diagram above, showing a hypothetical rock profile from two locations separated by a distance of 67 km. There are some differences between the rock layers at the two locations. Apart from layers D and L which are volcanic ash deposits, all other layers comprise sedimentary rock.

3. Assuming there has been no geologic activity (e.g. tilting or folding), state in which rock layer (A-O) you would find:

   (a) The youngest rocks at location 1: _____ (c) The youngest rocks at location 2: _____

   (b) The oldest rocks at location 1: _____ (d) The oldest rocks at location 2: _____

4. (a) State which layer at location 1 is of the same age as layer M at location 2: _____

   (b) Explain the reason for your answer above: _____

   _____

5. The rocks in layer H and O are sedimentary rocks. Explain why there are no visible fossils in these layers:

   _____

   _____

6. (a) State which layers present at location 1 are missing at location 2: _____

   (b) State which layers present at location 2 are missing at location 1: _____

7. Describe three methods of dating rocks: _____

   _____

   _____

8. Using radiometric dating, the trilobite fossil was determined to be approximately 375 million years old. The volcanic rock layer (D) was dated at 270 million years old, while rock layer B was dated at 80 million years old. Give the approximate **age range** (i.e. greater than, less than, or between given dates) of the rock layers listed below:

   (a) Layer A: _____ (d) Layer G: _____

   (b) Layer C: _____ (e) Layer L: _____

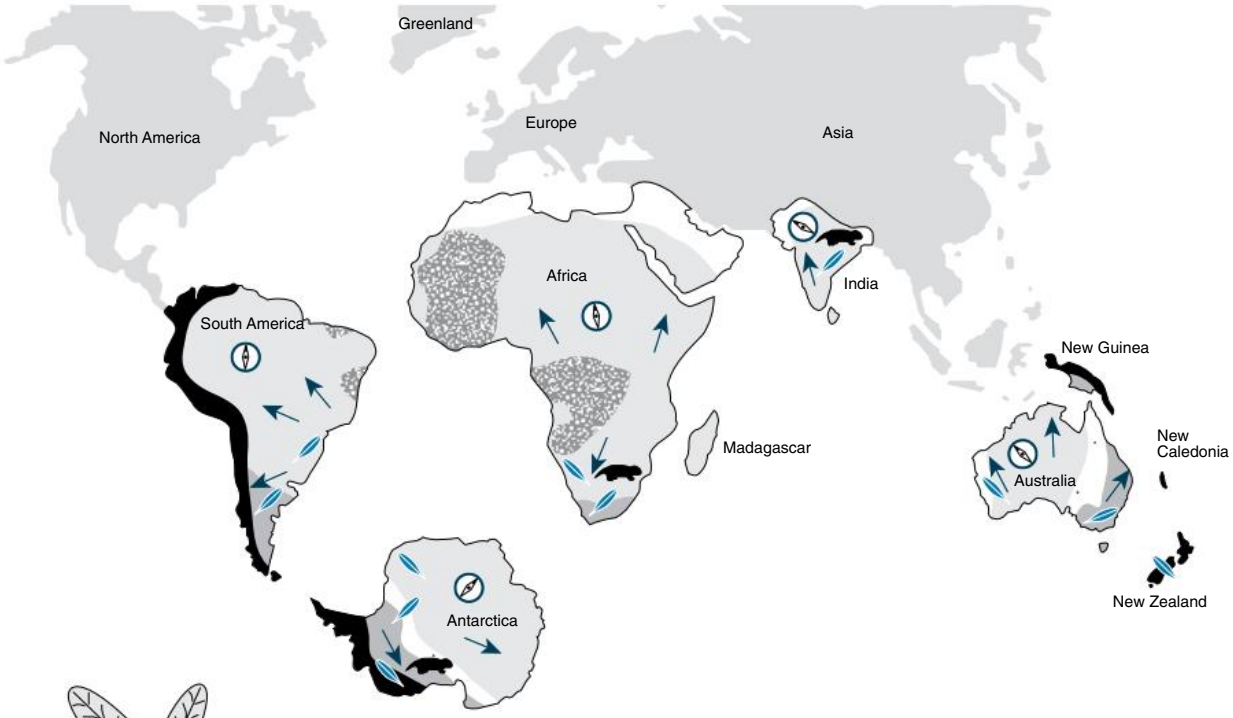
   (c) Layer E: _____ (f) Layer O: _____

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231 Continental Drift and Evolution

Key Idea: Fossils of related organisms found on separated continents can be explained by continental drift.

Continental drift (the movement of the Earth's continents relative to each other) is a measurable phenomenon; it has happened throughout Earth's history and continues today. Movements of up to 2-11 cm a year have been recorded between continents using laser technology. The movements of the Earth's 12 major crustal plates are driven by a geological process known as plate tectonics. Some continents are drifting apart while others are moving together. Many lines of evidence show that the modern continents were once joined together as 'supercontinents'. One supercontinent, **Gondwana**, was made up of the southern continents some 200 million years ago.





Glossopteris is a hardy plant that grew adjacent to the glacial ice sheets of Gondwana some 350-230 million years ago



Lystrosaurus is a primitive therapsid (mammal-like) reptile 1 m long, that was widely distributed throughout the southern continents about 240 million years ago.

Key

- Direction of ice sheet movement 350-230 million years ago
- Geomagnetic pole direction 150 million years ago
- Distribution of *Lystrosaurus*
- Distribution of *Glossopteris*
- Old Precambrian rocks (older than 650 mya)
- Precambrian basement rocks (650-570 mya)
- Early Palaeozoic folding (570-350 mya)
- Late Palaeozoic-Early Mesozoic folding (350-160 mya)
- Late Mesozoic folding (160-70 mya)

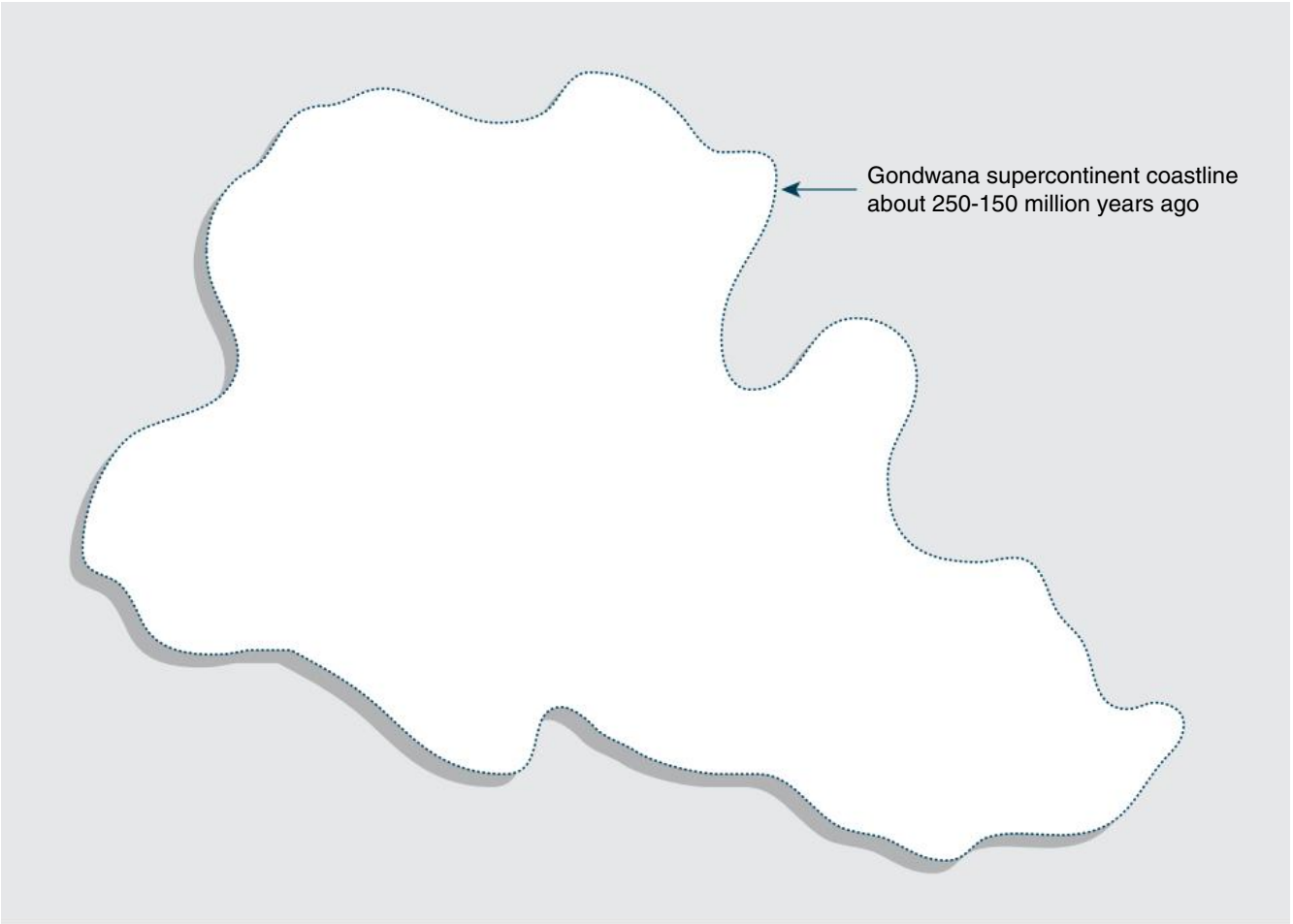
1. Name the modern landmasses (continents and large islands) that made up the supercontinent of Gondwana:

2. Cut out the southern continents on page 317 and arrange them to recreate the supercontinent of Gondwana. Take care to cut the shapes out close to the coastlines. When arranging them into the space showing the outline of Gondwana on page 316, take into account the following information:
   (a) The location of ancient rocks and periods of mountain folding during different geological ages.
   (b) The direction of ancient ice sheet movements.
   (c) The geomagnetic orientation of old rocks (the way that magnetic crystals are lined up in ancient rock gives an indication of the direction the magnetic pole was at the time the rock was formed).
   (d) The distribution of fossils of ancient species such as *Lystrosaurus* and *Glossopteris*.

3. Once you have positioned the modern continents into the pattern of the supercontinent, mark on the diagram:
   (a) The likely position of the South Pole 350-230 million years ago (as indicated by the movement of the ice sheets).
   (b) The likely position of the geomagnetic South Pole 150 million years ago (as indicated by ancient geomagnetism).

4. State what general deduction you can make about the position of the polar regions with respect to land masses:

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5. Fossils of *Lystrosaurus* are known from Antarctica, South Africa, India and Western China. With the modern continents in their present position, *Lystrosaurus* could have walked across dry land to get to China, Africa and India. It was not possible for it to walk to Antarctica, however. Explain the distribution of this ancient species in terms of continental drift:

6. The southern beech (*Nothofagus*) is found only in the southern hemisphere, in such places as New Caledonia, New Guinea, eastern Australia (including Tasmania), New Zealand, and southern South America. Fossils of southern beech trees have also been found in Antarctica. They have never been distributed in South Africa or India. The seeds of the southern beech trees are not readily dispersed by the wind and are rapidly killed by exposure to salt water.

   (a) Suggest a reason why *Nothofagus* is not found in Africa or India:

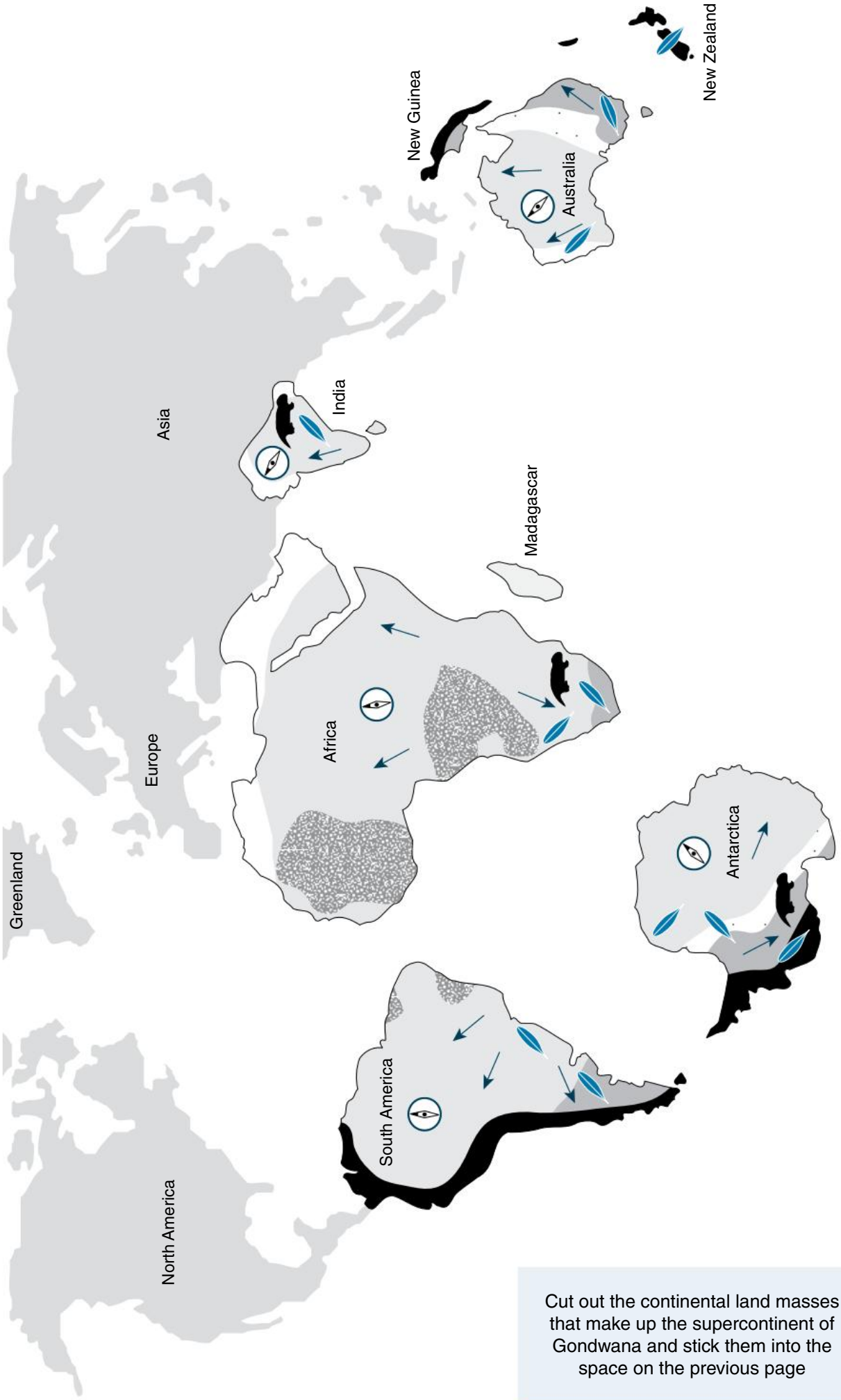
   (b) Use a colored pen to indicate the distribution of *Nothofagus* on the current world map (on the previous page) and on your completed map of Gondwana above.

   (c) State how the arrangement of the continents into Gondwana explains this distribution pattern:

7. The Atlantic Ocean is currently opening up at the rate of 2 cm per year. At this rate in the past, calculate how long it would have taken to reach its current extent, with the distance from Africa to South America being 2300 km (assume the rate of spreading has been constant):

8. Explain how continental drift provides evidence to support evolutionary theory:

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Cut out the continental land masses that make up the supercontinent of Gondwana and stick them into the space on the previous page

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QLD As required
VCE Unit 4 (2)
WA ATAR Unit 3
SA Theme E

Evolution

Key terms

adaptation
adaptive radiation
allopatric (allopatry)
analogous structure
background extinction rate
convergent evolution
divergent evolution (=cladogenesis)
evo-devo
evolution
extinction
hybrid
mass extinction
macroevolution
microevolution
migration
natural selection
parallel evolution
phyletic gradualism (=anagenesis)
phylogeny
population
postzygotic isolating mechanism
prezygotic isolating mechanism
reproductive isolation
shared derived characteristic
speciation
species
sympatric (sympatry)

Species and speciation

Learning aims and skills

		Activity number
☐	1 Explain what is meant by a biological species and describe the limitations of its definition, e.g. for extinct species and for asexually reproducing organisms.	232
☐	2 Recognise that phylogenetic species are defined on the basis of their shared ancestry, which is determined on the basis of shared derived characteristics.	233
☐	3 Describe and explain mechanisms of reproductive isolation, distinguishing between prezygotic and postzygotic reproductive isolating mechanisms.	234 235
☐	4 Explain allopatric speciation in terms of migration, isolation, and adaptation leading to reproductive (genetic) isolation of gene pools.	236
☐	5 Describe and explain sympatric speciation. Discuss the role of polyploidy in instant speciation events and in the development of some crops	237
☐	6 Describe an example of speciation in Australia, identifying the genus, species, and sub-species (if any), barriers to breeding between populations, the range of habitats occupied, any zones of sympatry, and recent range expansions.	238
☐	7 Recognise stages in species development, including reference to the reduction in gene flow as populations become increasingly isolated	239





Patterns of evolution

Learning aims and skills

		Activity number
☐	8 Describe patterns of species formation: phyletic gradualism, divergent evolution, adaptive radiation. Appreciate how evolutionary change over time has resulted in a great diversity among living organisms.	240-242 244
☐	9 Distinguish microevolutionary processes (natural selection, genetic drift, gene flow, and mutation) from large scale macroevolutionary patterns such as the formation of new species by adaptive radiation.	206 240
☐	10 Describe and explain convergent evolution. Explain how analogous structures may arise as a result of convergence. Distinguish between homologies, which are indicative of relatedness, and analogies, which are not.	243
☐	11 Distinguish between the punctuated equilibrium and gradualism models for the pace of evolutionary change. Discuss the evidence for each model.	245
☐	12 Describe the role of extinction in evolution. Distinguish clearly between background extinction rates and mass extinction. Identify the major mass extinctions and discuss the theories for their causes.	246



232 The Biological Species Concept

Key Idea: There is difficulty in using the biological species concept to define some species as many apparently separate species can interbreed to produce fertile offspring.

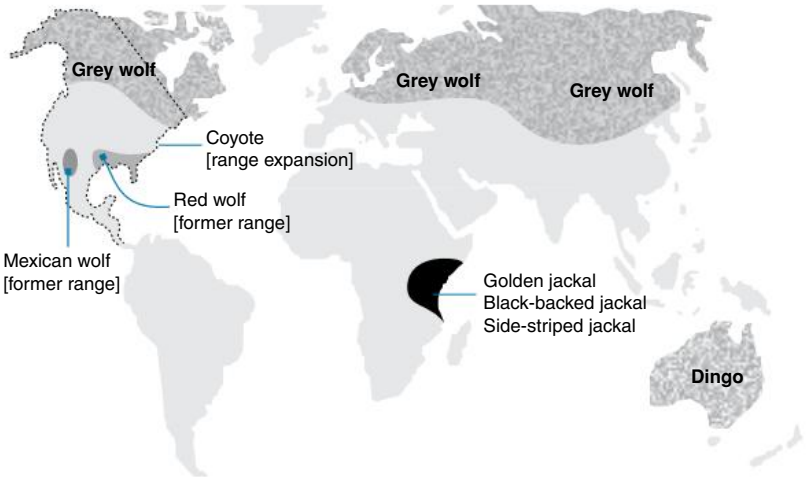
One of the best recognised definitions of a biological species is as "*a group of actually or potentially interbreeding natural populations that is reproductively isolated from other such groups*" (Ernst Mayr). However, the concept of a species is not always simple to apply. The concept is difficult to apply to fossil species and to asexual organisms. The occurrence of cryptic species and closely related species that interbreed to produce fertile hybrids (e.g. species of *Canis*), indicate that the boundaries of a species gene pool can be unclear.

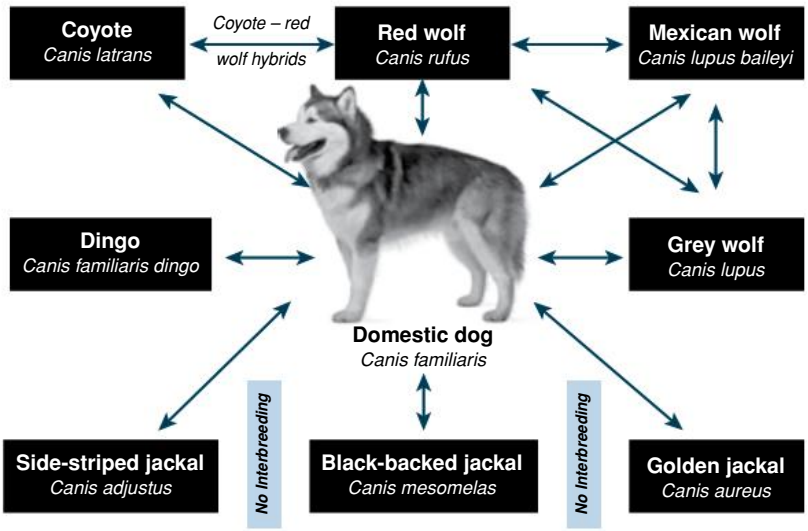
Geographical distribution of selected *Canis* species

The global distribution of most of the species of *Canis* (dogs and wolves) is shown on the map, right. The **grey wolf** inhabits the forests of North America, northern Europe, and Siberia. The **red wolf** and **Mexican wolf** (original distributions shown) were once distributed more widely, but are now extinct in the wild except for reintroduction efforts. In contrast, the coyote has expanded its original range and is now found throughout North and Central America. The range of the three **jackal** species overlap in the open savannah of Eastern Africa. The **dingo** is distributed throughout the Australian continent. Distribution of the domesticated **dog** is global as a result of the spread of human culture. The dog has been able to interbreed with all other members of the genus listed here to form fertile hybrids.

Interbreeding between *Canis* species

The *Canis* species illustrate problems with the traditional species concept. The domesticated dog is able to breed with other members of the same genus to produce fertile hybrids. Red wolves, grey wolves, Mexican wolves, and coyotes are all capable of interbreeding to produce fertile hybrids. Red wolves are very rare, and it is possible that hybridisation with coyotes has been a factor in their decline. By contrast, the ranges of the three distinct species of jackal overlap in the Serengeti of Eastern Africa. These animals are highly territorial, but simply ignore members of the other jackal species and no interbreeding takes place.





1. What type of barrier prevents interbreeding between the three jackal species? _____

2. Describe the barrier preventing interbreeding between the dingo and other *Canis* species (apart from the dog):

_____

3. Describe a possible contributing factor to the occurrence of interbreeding between the coyote and red wolf:

_____

4. The grey wolf is a widely distributed species. Explain why the North American and the northern European and Siberian populations are considered to be the same species:

_____

_____

5. Explain what you understand by the term species, identifying examples where the definition is problematic:

_____

_____

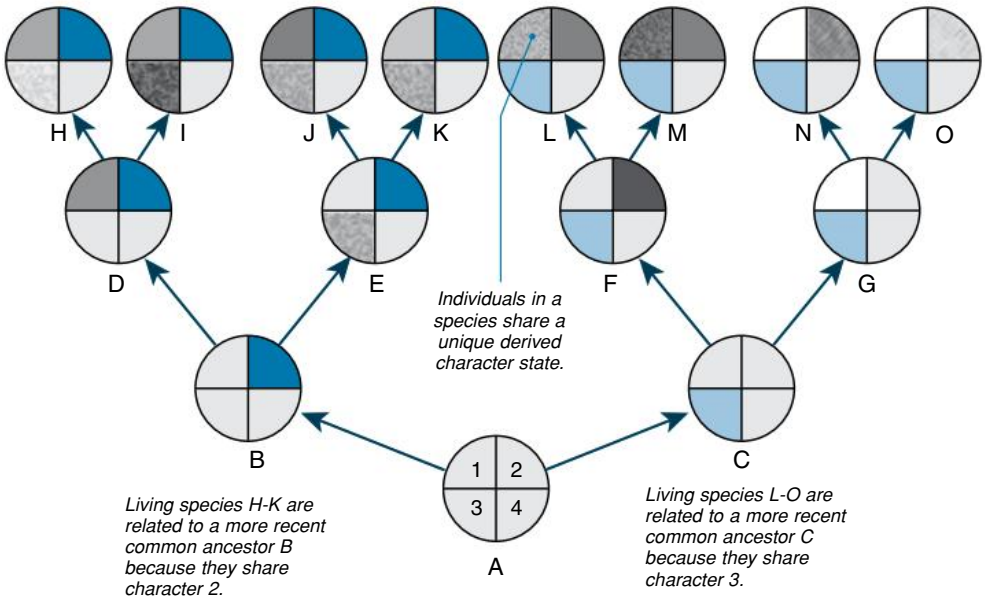
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233 The Phylogenetic Species Concept

Key Idea: Phylogenetic species are determined on the basis of shared derived characteristics.

Although the biological species concept is useful, there are many situations in which it is difficult to apply, e.g. for asexual populations (including bacteria) or extinct organisms. In such situations, the **phylogenetic species concept** (PSC) can be more useful. It not is reliant on the criterion of successful interbreeding and can be applied to asexually or sexually reproducing organisms, and to extinct organisms. Phylogenetic species are defined on the basis of their shared evolutionary ancestry, which is determined on the basis of **shared derived characteristics**, which may be morphological, especially for higher taxonomic ranks, or biochemical (e.g. DNA differences). The PSC defines a species as the smallest group that all share a derived character state. While the PSC solves some difficulties, it creates others. For example, the ability to distinguish genetically distinct but morphologically identical cryptic species on the basis of DNA analyses can lead to a proliferation of existing species that is not helpful in establishing a phylogeny.



This simplified phylogenetic tree traces four characters among 15 species (8 present and 7 ancestral). The 8 modern species (species H-O) share a character (4) derived from a distant common ancestor (A). Although the primitive character unites all 8 species, the branching of the tree is based on characters derived from the ancestral ones. Classification on the basis of shared derived characters defines the species as the smallest group diagnosable by a unique combination of characters. If large numbers of characters are included in the analysis, it is easy to see how this method results in a proliferation of species that may or may not be meaningful. Under the PSC model, there are no subspecies; either a population is a phylogenetic species or it is not taxonomically distinguishable.



Tree sparrows (*P. montanus*) are ~10% smaller than the similar house sparrow but the two species hybridise freely.



House sparrows (*P. domesticus*) are widespread with many intermediate "subspecies" of unknown status.



Mallards are infamous for their ability to hybridise freely with a large number of other duck "species".

True sparrows all belong to the genus *Passer*. There are a large number of species distinguished on the basis of song, plumage, and size. A vestigial dorsal outer primary feather and an extra bone in the tongue are ancestral characters. Many populations are not good biological species in that they hybridise freely to produce fertile offspring. A similar situation exists within the genus *Anas* of dabbling ducks (which includes the mallards). Many birds are best described using the PSC rather than the BSC.

1. (a) Explain the basis by which species are assigned under the PSC: _____

   _____

   _____

   (b) Describe one problem with the use of the PSC: _____

   _____

   (c) Describe situations where the use of the PSC might be more appropriate than the BSC: _____

   _____

   _____

2. Suggest how genetic techniques could be used to elucidate the phylogeny of a cluster of related phylogenetic species:

   _____

   _____

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LINK 234 233 WEB KNOW

234 Isolation and Species Formation

Key Idea: Ecological and geographical isolation are important in separating populations prior to reproductive isolation.

Isolating mechanisms are barriers to successful interbreeding between species. **Reproductive isolation** is fundamental to the biological species concept, which defines a species by its inability to breed with other species to produce fertile offspring. **Geographical barriers** are not regarded as reproductive isolating mechanisms because they are not part of the species' biology, although they are often a necessary precursor to reproductive isolation in sexually reproducing populations. Ecological isolating mechanisms are those that isolate gene pools on the basis of ecological preferences, e.g. habitat selection. Although ecological and geographical isolation are sometimes confused, they are quite distinct, as ecological isolation involves a component of the species biology.

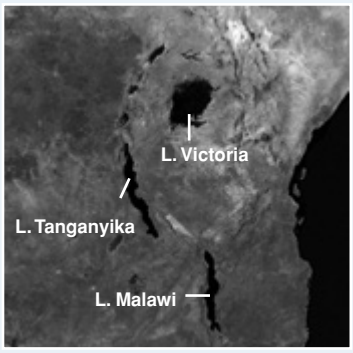
Geographical Isolation

Geographical isolation describes the isolation of a species population (gene pool) by some kind of physical barrier, for example, mountain range, water body, isthmus, desert, or ice sheet. Geographical isolation is a frequent first step in the subsequent reproductive isolation of a species. For example, geological changes to the lake basins has been instrumental in the subsequent proliferation of cichlid fish species in the rift lakes of East Africa (right). Similarly, many Galapagos Island species (e.g. iguanas, finches) are now quite distinct from the Central and South American species from which they arose after isolation from the mainland.

Ecological (Habitat) Isolation

Ecological isolation describes the existence of a **prezygotic reproductive barrier** between two species (or sub-species) as a result of them occupying or breeding in different habitats within the same general geographical area. Ecological isolation includes small scale differences (e.g. ground or tree dwelling) and broad differences (e.g. desert vs grasslands). The red-browed and brown **treecreepers** (*Climacteris* spp.) are sympatric in south-eastern Australia and both species feed largely on ants. However the brown spends most of its time foraging on the ground or on fallen logs while the red-browed forages almost entirely in the trees. Ecological isolation often follows geographical isolation, but in many cases the geographical barriers may remain in part. For example, five species of **antelope squirrels** occupy different habitat ranges throughout the southwestern United States and northern Mexico, a region divided in part by the Grand Canyon. The white tailed antelope squirrel is widely distributed in desert areas to the north and south of the canyon, while the smaller, more specialised Harris' antelope squirrel has a much more limited range only to the south in southern Arizona. The Grand Canyon still functions as a barrier to dispersal but the species are now ecologically isolated as well.

Geographical and Ecological Isolation of Species



NASA Earth Observatory



istock


Both photos: Aviceda

Red-browed treecreeper



Brown treecreeper


UtahCamera

White-tailed antelope squirrel


istock

The Grand Canyon - a massive rift in the Colorado Plateau


Photo: Allan and Elaine Wilson

Harris' antelope squirrel

1. Describe the role of isolating mechanisms in maintaining the integrity of a species: _____

2. (a) Why is geographical isolation not regarded as a reproductive isolating mechanism? _____

   (b) Explain why, despite this, it often precedes reproductive isolation: _____

3. Distinguish between geographical and ecological isolation: _____

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235 Reproductive Isolation

Key Idea: Reproductive isolating mechanisms prevent interbreeding between species. They include prezygotic and postzygotic isolating mechanisms.

Reproductive isolation is a defining feature of biological species. Any mechanism that prevents two species from producing viable, fertile hybrids contributes to reproductive isolation. Single barriers to gene flow (such as geographical barriers) are usually insufficient to isolate a gene pool, so most species commonly have more than one type of barrier. Most reproductive isolating mechanisms (RIMs) are prezygotic and operate before fertilisation. Postzyotic RIMs are important in maintaining the integrity of closely related species.

Prezygotic Isolating Mechanisms

Temporal Isolation

Individuals from different species do not mate because they are active during different times of the day or in different seasons. Plants flower at different times of the year or at different times of the day to avoid hybridisation (e.g. species of the orchid genus *Dendrobium* occupy the same location but flower on different days). Closely related animal species may have different breeding seasons or periods of emergence. Species of **periodical cicadas** (*Magicicada*) in a particular region are developmentally synchronised, despite very long life cycles. Once their underground period of development (13 or 17 years depending on the species) is over, the entire population emerges at much the same time to breed.

Gamete Isolation

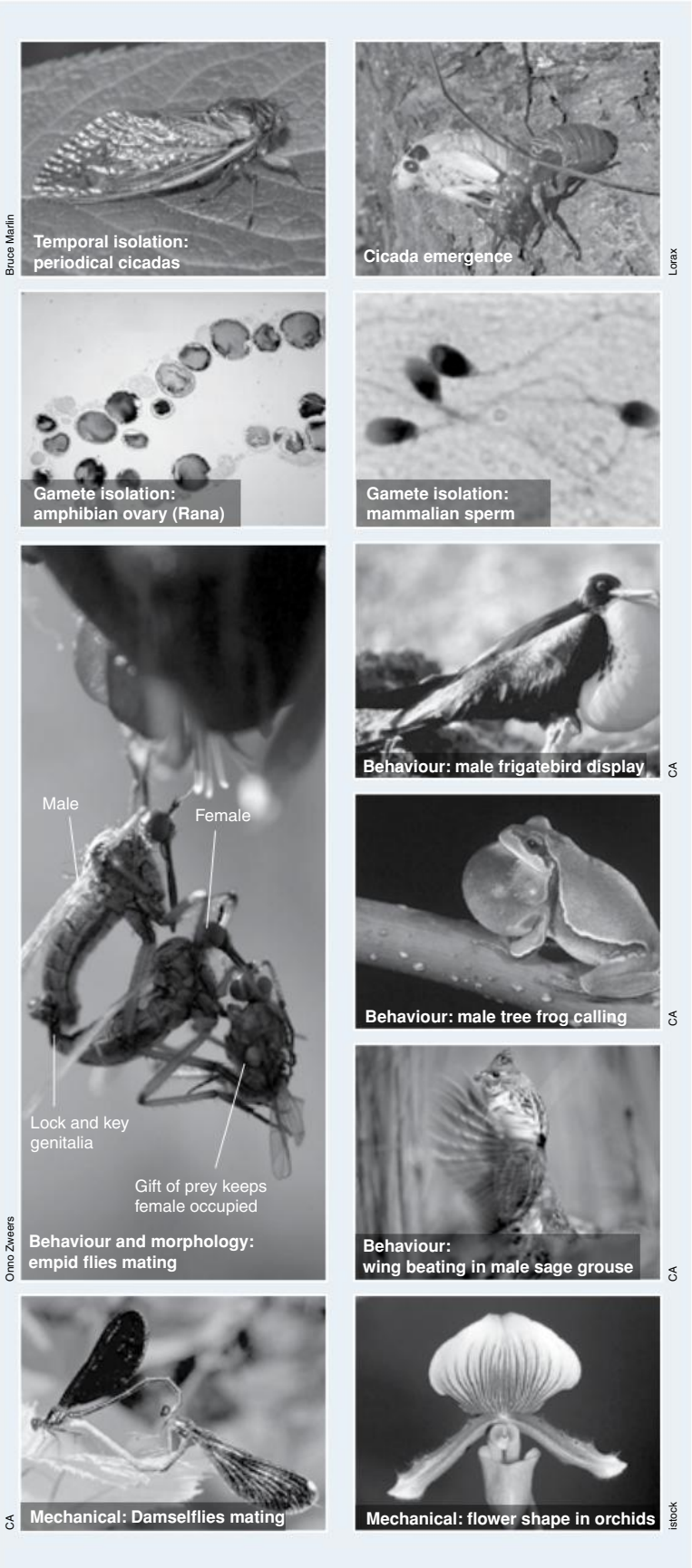
The gametes from different species are often incompatible, so even if they meet they do not survive. Where fertilisation is internal, the sperm may not survive in the reproductive tract of another species. If the sperm does survive and reach the ovum, chemical differences in the gametes prevent fertilisation. Gamete isolation is particularly important in aquatic environments where the gametes are released into the water and fertilised externally, such as in reproduction in frogs. Chemical recognition is also used by flowering plants to recognise pollen from the same species.

Behavioural Isolation

Behavioural isolation operates through differences in species courtship behaviours. Courtship is a necessary prelude to mating in many species and courtship behaviours are species specific. Mates of the same species are attracted with distinctive dances, vocalisations, and body language. Courtship behaviours are not easily misinterpreted and will be unrecognised and ignored by individuals of another species. Birds exhibit a remarkable range of courtship displays. The use of song is widespread but ritualised movements, including nest building, are also common. For example, the elaborate courtship bowers of bowerbirds are well known, and Galápagos frigatebirds have an elaborate display in which they inflate a bright red gular pouch (right). Amongst insects, empid flies have some of the most elaborate of courtship displays. They are aggressive hunters so ritualised behaviour involving presentation of a prey item facilitates mating. The sexual organs of the flies are also like a lock-and-key, providing mechanical reproductive isolation as well (see below).

Mechanical (morphological) Isolation

Structural differences (incompatibility) in the anatomy of reproductive organs prevents sperm transfer between individuals of different species. This is an important isolating mechanism preventing breeding between closely related species of arthropods. Many flowering plants have coevolved with their animal pollinators and have flowers structures to allow only that insect access. Structural differences in the flowers and pollen of different plant species prevents cross breeding because pollen transfer is restricted to specific pollinators and the pollen itself must be species compatible.



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Postzygotic Isolating Mechanisms

Hybrid sterility

Even if two species mate and produce hybrid offspring that are vigorous, the species are still reproductively isolated if the hybrids are sterile (genes cannot flow from one species' gene pool to the other). Such cases are common among the horse family (such as the zebra and donkey shown on the right). One cause of this sterility is the failure of meiosis to produce normal gametes in the hybrid. This can occur if the chromosomes of the two parents are different in number or structure (see the "**zebronkey**" karyotype on the right). The **mule**, a cross between a donkey stallion and a horse mare, is also an example of **hybrid vigour** (they are robust) as well as **hybrid sterility**. Female mules sometimes produce viable eggs but males are infertile.

Hybrid inviability

Mating between individuals of two species may produce a zygote, but genetic incompatibility may stop development of the zygote. Fertilised eggs often fail to divide because of mis-matched chromosome numbers from each gamete. Very occasionally, the hybrid zygote will complete embryonic development but will not survive for long. For example, although sheep and goats seem similar and can be mated together, they belong to different genera. Any offspring of a sheep-goat pairing is generally stillborn.

Hybrid breakdown

Hybrid breakdown is common feature of some plant hybrids. The first generation (F_1) may be fertile, but the second generation (F_2) are infertile or inviable. Examples include hybrids between cotton species (near right), species within the genus *Populus*, and strains of the cultivated rice *Oryza* (far right)



1. In the following examples, classify the reproductive isolating mechanism as either **prezygotic** or **postzygotic** and describe the mechanisms by which the isolation is achieved (e.g. structrual isolation, hybrid sterility etc.):

   (a) Some different cotton species can produce fertile hybrids, but breakdown of the hybrid occurs in the next generation when the offspring of the hybrid die in their seeds or grow into defective plants:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

   (b) Many plants have unique arrangements of their floral parts that stops transfer of pollen between plants:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

   (c) Two skunk species do not mate despite having habitats that overlap because they mate at different times of the year:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

   (d) Several species of the frog genus *Rana*, live in the same regions and habitats, where they may occasionally hybridise. The hybrids generally do not complete development, and those that do are weak and do not survive long:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

2. Postzygotic isolating mechanisms are said to reinforce prezygotic ones. Explain why this is the case:

_____

_____

_____

_____

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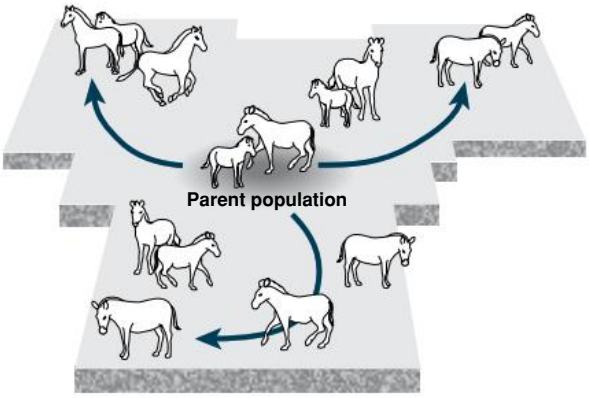
236 Allopatric Speciation

Key Idea: Allopatric speciation is the genetic divergence of a population after it becomes subdivided and isolated.

Allopatric speciation refers to divergence of a species after it is subdivided into geographically isolated populations. It is probably the most common mechanism by which new species arise and has certainly been important in regions where there have been cycles of geographical fragmentation. For example, during glacial periods, Tasmania and New Guinea formed a single land mass with the Australian continent, but during interglacial periods, these connections disappeared.

Stage 1: Moving into new environments

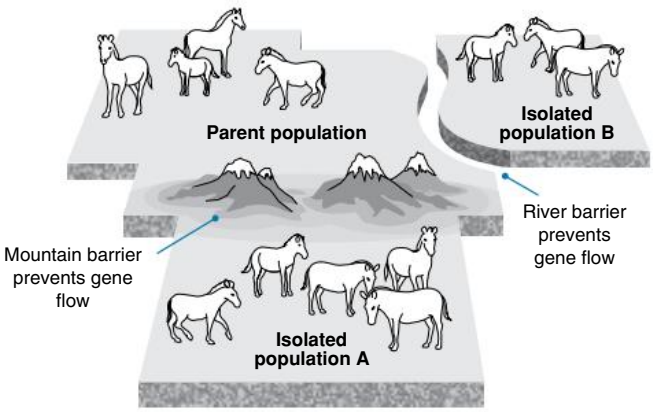
There are times when the range of a species expands for a variety of different reasons. A single population in a relatively homogeneous environment will move into new regions of their environment when they are subjected to intense competition (whether it is interspecific or intraspecific). The most severe form of competition is between members of the same species since they are competing for identical resources in the habitat. In the diagram on the right there is a 'parent population' of a single species with a common gene pool with regular 'gene flow' (theoretically any individual has access to all members of the opposite sex for mating purposes).



Stage 2: Geographical isolation

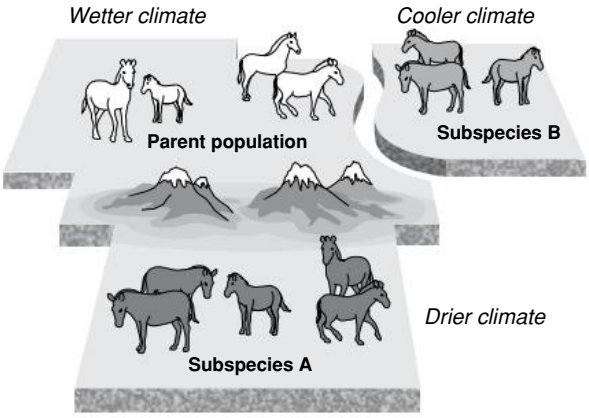
Isolation of parts of the population may occur due to the formation of **physical barriers**, such as mountains, deserts, or stretches of water. These barriers may cut off those parts of the population that are at the extremes of the range and gene flow is prevented or rare. The rise and fall of the sea level has been particularly important in functioning as an isolating mechanism. Climatic change can leave 'islands' of habitat separated by large inhospitable zones that the species cannot traverse.

Example: In mountainous regions, alpine species can populate extensive areas of habitat during cool climatic periods. During warmer periods, they may become isolated because their habitat is reduced to 'islands' of high ground surrounded by inhospitable lowland habitat.



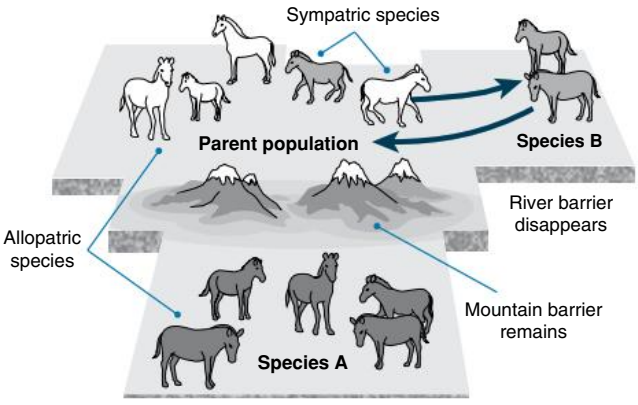
Stage 3: Different selection pressures

The isolated populations (A and B) may be subjected to quite different selection pressures. These will favour individuals with traits that suit each particular environment. For example, population A will be subjected to selection pressures that relate to drier conditions. This will favour those individuals with phenotypes (and therefore genotypes) that are better suited to dry conditions. They may for instance have a better ability to conserve water. This would result in improved health, allowing better disease resistance and greater reproductive performance (i.e. more of their offspring survive). Finally, as allele frequencies for certain genes change, the population takes on the status of a subspecies. Reproductive isolation is not yet established but the **subspecies** are significantly different genetically from other related populations.



Stage 4: Reproductive isolation

The separated populations (isolated subspecies) undergo genetic and behavioural changes. These ensure that the gene pool of each population remains isolated and 'undiluted' by genes from other populations, even if the two populations should be able to remix (due to the removal of the geographical barrier). Gene flow does not occur. The arrows (diagram, right) indicate the zone of overlap between two species after Species B has moved back into the range inhabited by the parent population. Closely-related species whose distribution overlaps are said to be **sympatric species**. Those that remain geographically isolated are called **allopatric species**.



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1. Why do some animals, given the opportunity, move into new environments? _____

2. Plants are unable to move. How might plants disperse to new environments? _____

3. Describe the amount of **gene flow** within a parent population prior to and during the expansion of a species' range:

4. Explain how cycles of climate change can cause large changes in **sea level** (up to 200 m):

5. (a) What kinds of **physical barriers** could isolate different parts of the same population? _____

   (b) How might emigration achieve the same effect as geographical isolation? _____

6. (a) How might **selection pressures** differ for a population that becomes isolated from the parent population?

   (b) Describe the general effect of the change in selection pressures on the **allele frequencies** of the isolated gene pool:

7. Explain how reproductive isolation could develop in geographically separated populations (see previous pages):

8. What is the difference between an allopatric and sympatric species? _____

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237 Sympatric Speciation

Key Idea: Sympatric speciation is speciation occurring in the absence of physical barriers between gene pools.

Sympatric speciation is rarer than allopatric speciation, although it is not uncommon in plants, which can form polyploids. There are two situations in which sympatric speciation is thought to occur. These are described below.

Speciation Through Niche Differentiation

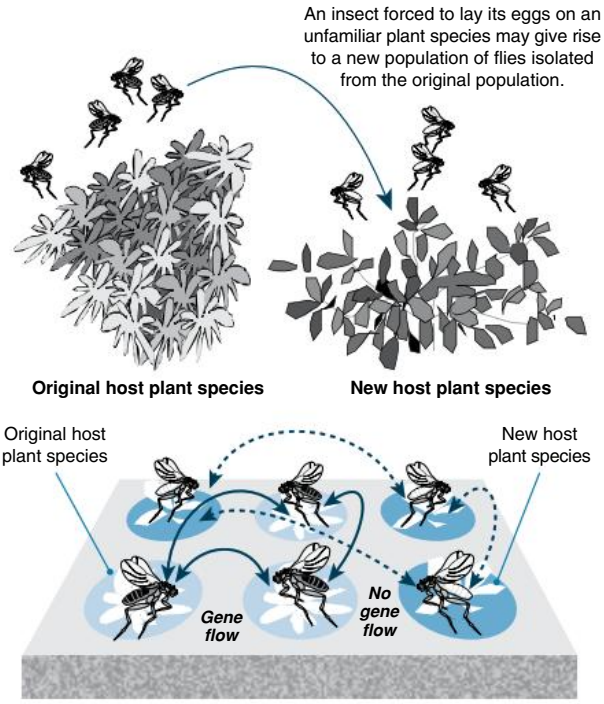
Niche isolation

In a heterogeneous environment (one that is not the same everywhere), a population exists within a diverse collection of **microhabitats**. Some organisms prefer to occupy one particular type of 'microhabitat' most of the time, only rarely coming in contact with fellow organisms that prefer other microhabitats. Some organisms become so dependent on the resources offered by their particular microhabitat that they never meet up with their counterparts in different microhabitats.

Reproductive isolation

Finally, the individual groups have remained genetically isolated for so long because of their microhabitat preferences, that they have become reproductively isolated. They have become new species that have developed subtle differences in behaviour, structure, and physiology. Gene flow (via sexual reproduction) is limited to organisms that share a similar microhabitat preference (as shown in the diagram on the right).

Example: Some beetles prefer to find plants identical to the species they grew up on, when it is time for them to lay eggs. Individual beetles of the same species have different preferences.



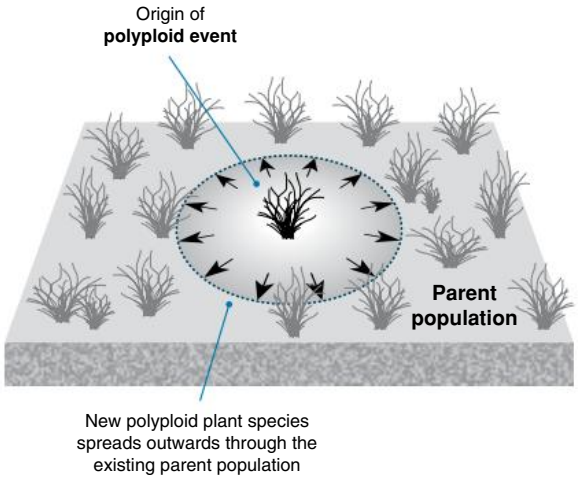
Instant Speciation by Polyploidy

Polyploidy may result in the formation of a new species without isolation from the parent species. This event, occurring during meiosis, produces sudden reproductive isolation for the new group. Because the sex-determining mechanism is disturbed, animals are rarely able to achieve new species status this way (they are effectively sterile, e.g. tetraploid XXXX). Many plants, on the other hand, are able to reproduce vegetatively, or carry out self pollination. This ability to reproduce on their own enables such polyploid plants to produce a breeding population.

Speciation by allopolyploidy

This type of polyploidy usually arises from the doubling of chromosomes in a hybrid between two different species. The doubling often makes the hybrid fertile.

Examples: Modern wheat. Swedes are polyploid species formed from a hybrid between a type of cabbage and a type of turnip.



1. Explain what is meant by **sympatric speciation** (do not confuse this with sympatric species):

2. Explain how **polyploidy** can result in the formation of a new species:

3. Identify an example of a species that has been formed by polyploidy:

4. Explain how **niche differentiation** can result in the formation of a new species:

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238 Speciation in Australia

Key Idea: Geographic barriers presented by inhospitable habitat are contributing to speciation in Australian treecreepers.

The geographical barriers isolating populations on continents are often very different to those isolating island populations. In Australia, geographical barriers exist in the form of regions of inhospitable habitat. These create regions of preferred habitat cut off from one another. The species and subspecies of Australian treecreepers, *Climacteris*, are distinguishable by variations in the colour patterns of their plumage. Their distribution is restricted to savannah woodland and a distinctive form is associated with each of the major woodland areas (below). The populations probably evolved from a single common ancestor, isolated by habitat changes and then unable to expand their individual distributions beyond regions of unsuitable dry habitat. Subsequently, the distribution of two of the treecreeper species has undergone a secondary range expansion (arrows), where they have extended their range beyond their region of origin, into new habitat.

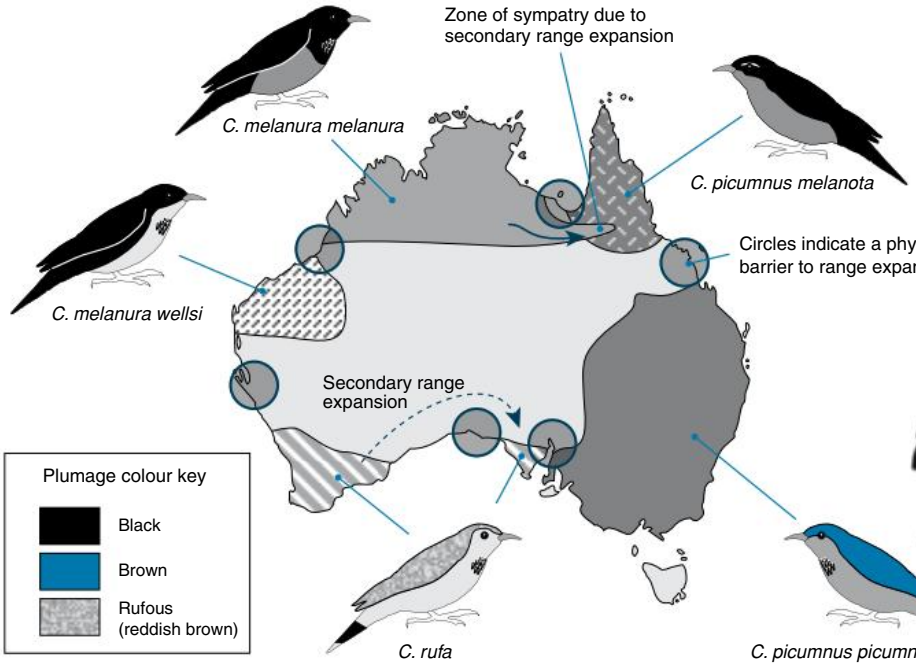




Photo: Aviceda

The Australian treecreeper
Genus *Climacteris*

1. (a) How many **species** are illustrated above? Explain your answer: _____

   _____

   (b) Describe the distribution of these treecreeper populations in Australia: _____

   _____

   _____

2. Explain why there are no treecreeper populations in the central region of Australia: _____

   _____

3. Two species in the NE of Australia, *C. melanura melanura* and *C. picumnus melanota*, exhibit sympatric distribution.

   (a) What is meant by the area of sympatry in this context? _____

   _____

   (b) What mechanisms are most likely to prevent interbreeding between these two species? _____

   _____

4. What is meant by **secondary range expansion** in the two populations above: _____

   _____

5. Describe the physical barriers that have prevented the neighbouring populations from mixing (in all but one case):

   _____

6. Predict a likely outcome to the distribution of these species, should the climate change to produce more coastal rainfall:

   _____

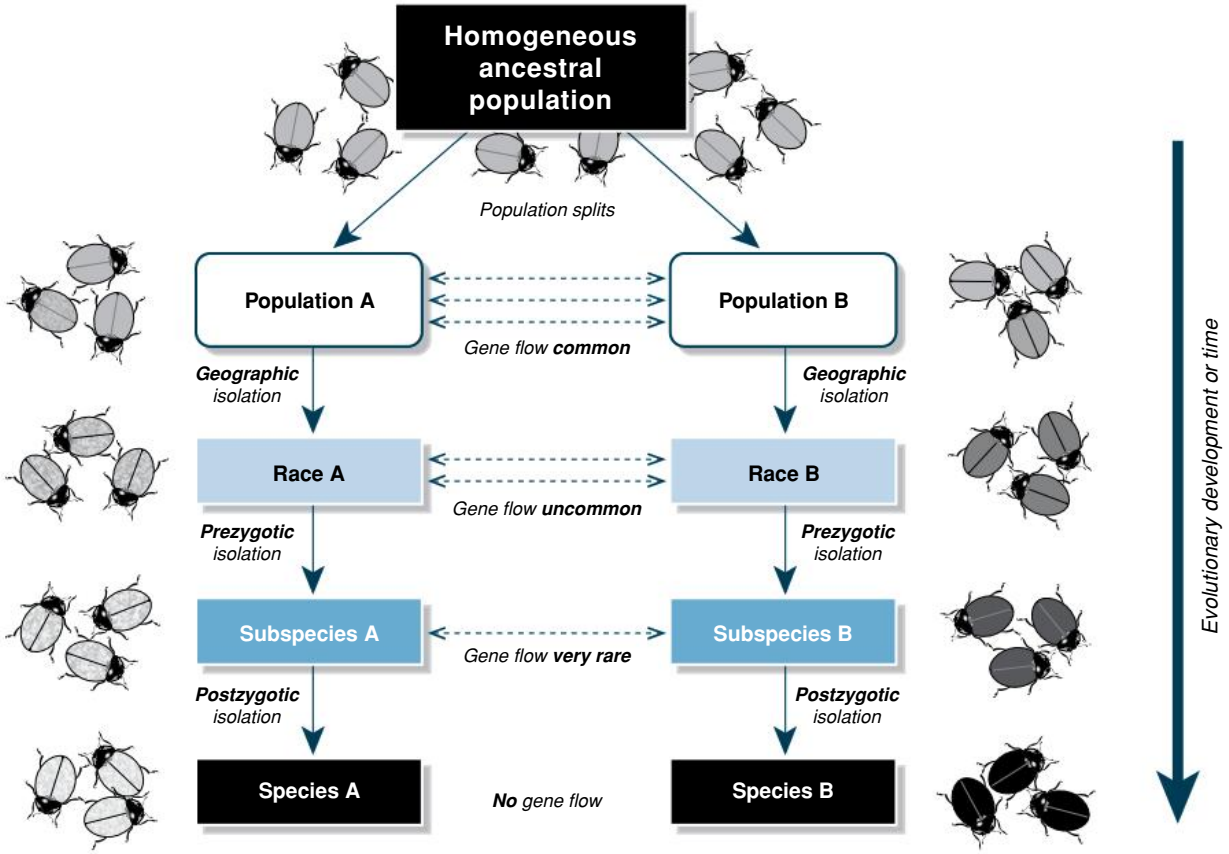
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239 Stages in Species Development

Key Idea: Species may develop in stages marked by increasing isolation of diverging gene pools. Physical separation is followed by prezygotic isolation (e.g. behaviour) and finally postzygotic isolation (e.g. hybrid breakdown).

The diagram below shows a possible sequence of events in the origin of two new species from an ancestral population. Over time, the genetic differences between two populations increase and the populations become increasingly isolated from each other. The isolation of the two gene pools may begin with a geographical barrier. This may be followed by prezygotic mechanisms, such as behavioural differences, which reduce gene flow between the gene pools. Longer isolation may lead to postzygotic mechanisms. As the two gene pools become increasingly isolated and divergent, they are progressively labelled: populations, races, and subspecies. Finally, they each attain species status.



1. What happens to the amount of gene flow between two diverging populations as they gradually attain species status?

2. Early human populations about 500,000 years ago were scattered across the continents of Africa, Europe, and Asia. This was a time of many regional variants, collectively called archaic *Homo sapiens* (or *Homo heidelbergensis*). The fossil skulls from different regions showed odd mixtures of characteristics, some modern and some considered 'primitive'. These regional populations are generally given the status of subspecies. Suggest reasons why gene flow may have been rare between these populations, but still occasionally took place:

3. The Australian race of Pacific black duck and the introduced mallard duck (from the Northern hemisphere) are undergoing 'species breakdown'. These two closely related species can interbreed to form hybrids.

   (a) Describe the factor preventing the two species interbreeding before the introduction of the mallards to Australia:

   (b) Describe the factor that may be deterring some of the ducks from interbreeding with the other species:

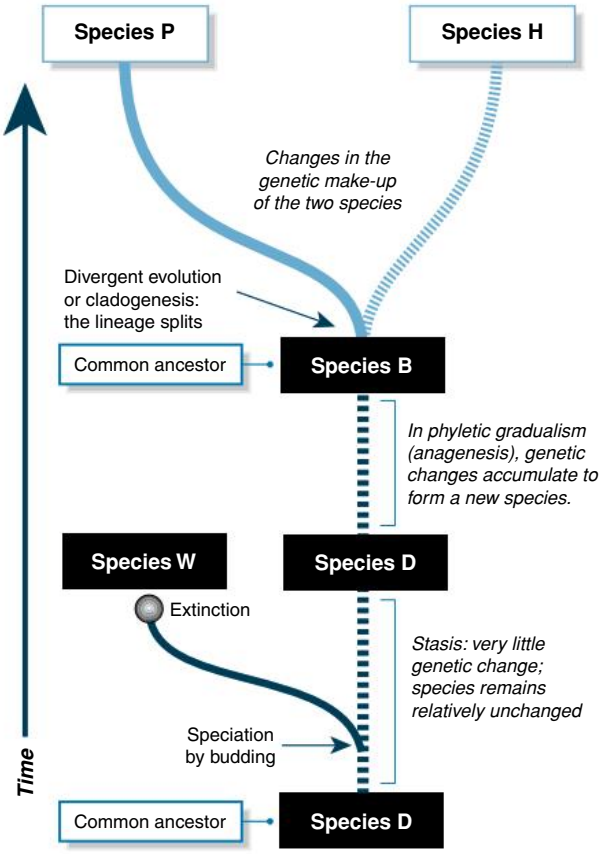
   (c) Why do these hybrids threaten the species status of native duck species?

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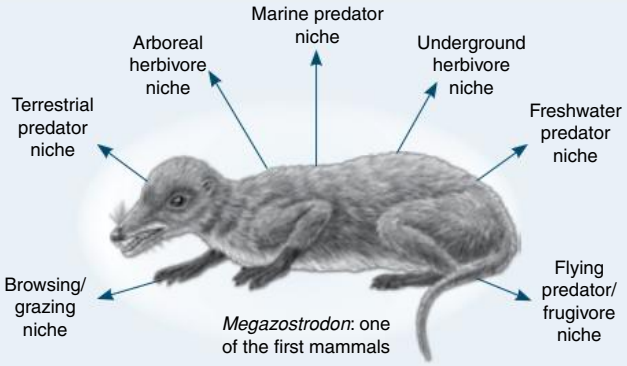
240 Patterns of Evolution

Key Idea: Populations moving into a new environment may diverge from their common ancestor and form new species.

The diversification of an ancestral group into two or more species in different habitats is called **divergent evolution**. This process is shown below, where two species have diverged from a **common ancestor**. Divergence is common in evolution. When divergent evolution involves the formation of a large number of species to occupy different niches, this is called an **adaptive radiation**. The example below (right) describes the radiation of the mammals after the extinction of the dinosaurs made new niches available. Note that the evolution of species may not necessarily involve branching. A species may accumulate genetic changes that, over time, result in the emergence of what can be recognised as a different species. This is known as **phyletic gradualism** (also called sequential evolution or anagenesis).



Mammalian Adaptive Radiation



The earliest true mammals evolved about 195 million years ago, long before they underwent their major adaptive radiation some 65-50 million years ago. These ancestors to the modern forms were very small (12 cm), many were nocturnal and fed on insects and other invertebrates. *Megazostrodon* (above) is a typical example. This shrew-like animal is known from fossil remains in South Africa and first appeared in the Early Jurassic period (about 195 million years ago).

It was climatic change as well as the extinction of the dinosaurs (and their related forms) that suddenly left many niches vacant for exploitation by such adaptable 'generalists'. All modern mammal orders developed very quickly and early.

1. In the hypothetical example of divergent evolution illustrated above, left:

   (a) Describe the type of evolution that produced species B from species D: _____

   (b) Describe the type of evolution that produced species P and H from species B: _____

   (c) List all species that evolved from: **Common ancestor D**: _____ **Common ancestor B**: _____

   (d) Explain why species B, P, and H all possess a physical trait not found in species D or W: _____

   _____

2. (a) Explain the distinction between **divergence** and **adaptive radiation**: _____

   _____

   _____

   (b) What are the differences between **sequential evolution** and **divergent evolution**? _____

   _____

   _____

   _____

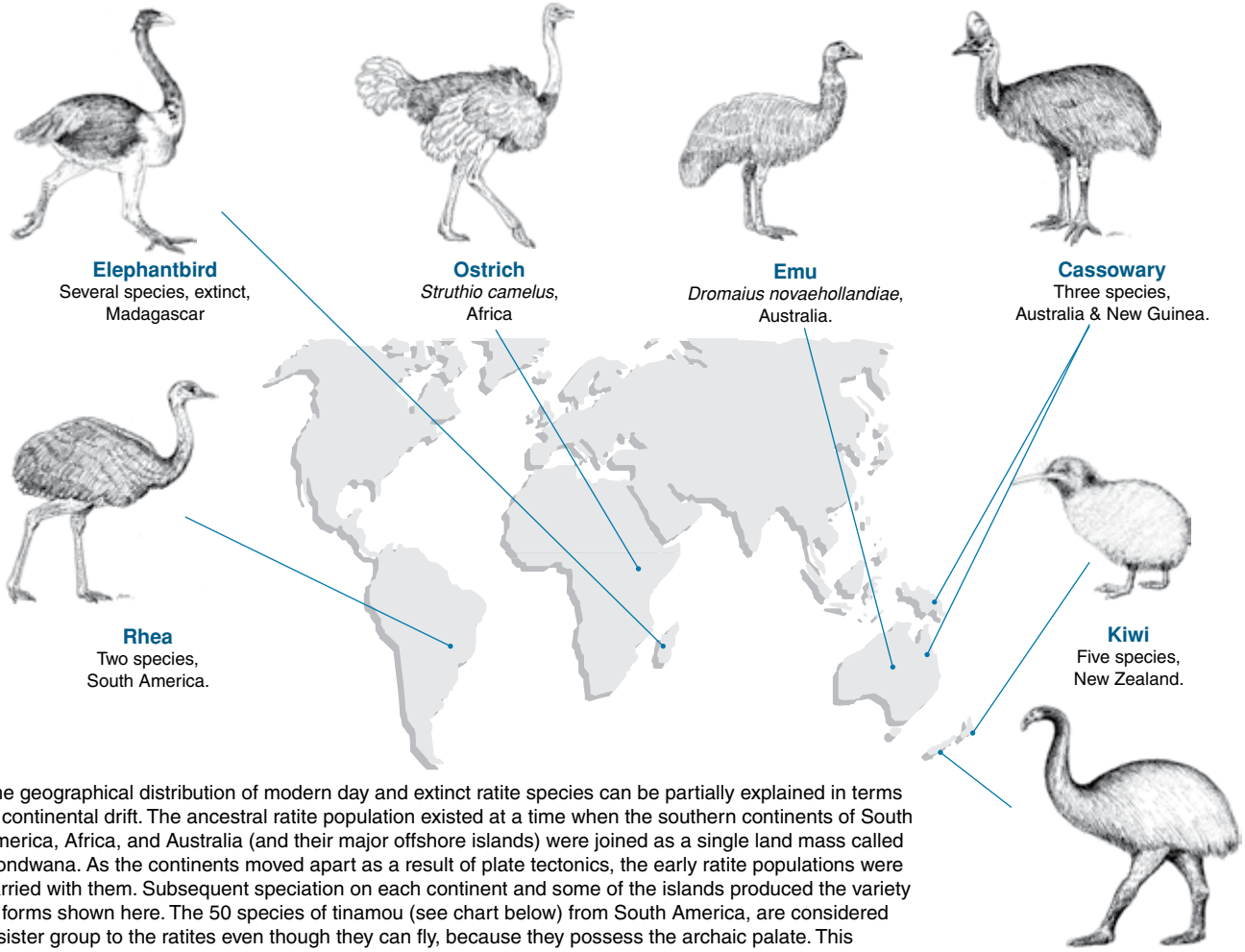
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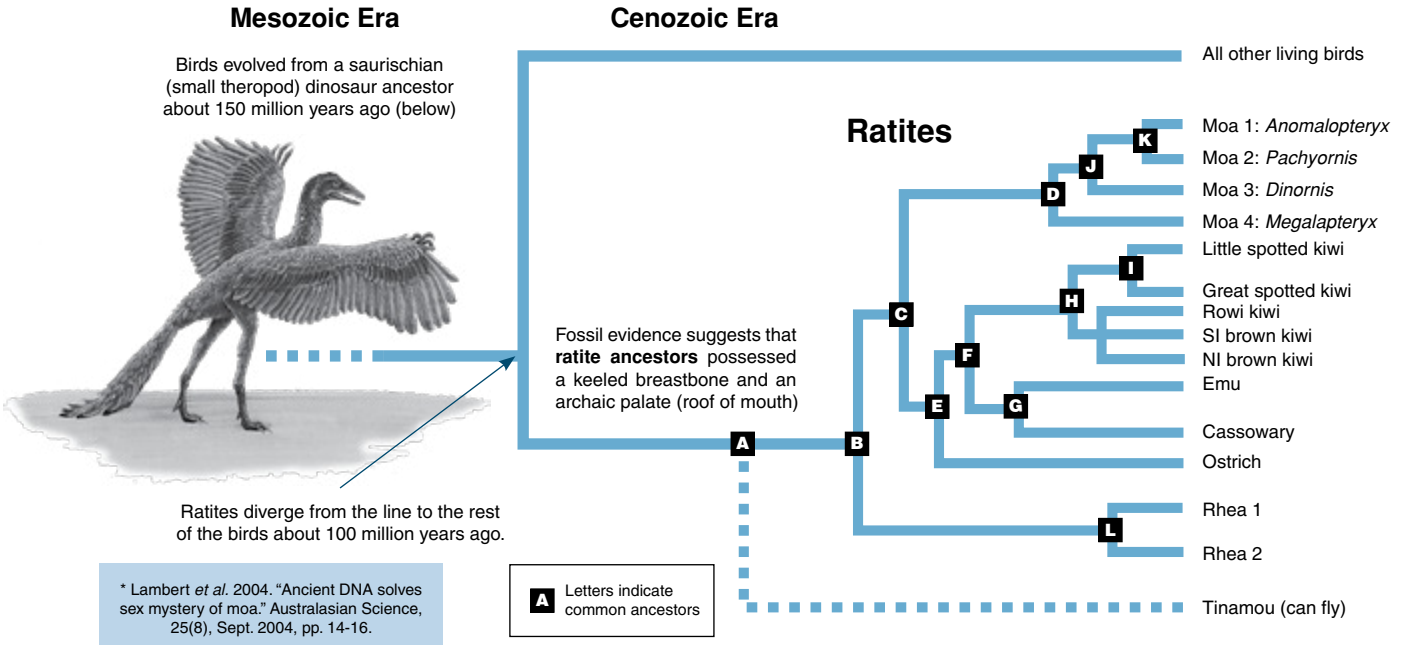
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241 Adaptive Radiation in Ratites

Key Idea: The ratites are a monophyletic group of birds (descended from a single common ancestor) that lost the power of flight very early on in their evolutionary development. Ratites are flightless birds that possess two features that distinguish them from other birds; a flat breastbone (instead of the more usual keeled shape) and a primitive palate (roof to the mouth). Fossil evidence indicates that the ancestors of ratites were flying birds living about 80 million years ago. These ancestors also had a primitive palate, but they possessed a keeled breastbone. Flightlessness in itself is not unique to ratites; there are other birds that have lost the power of flight, particularly on remote, predator-free islands. All ratites have powerful legs, and many, such as the emu, can run very quickly.



The geographical distribution of modern day and extinct ratite species can be partially explained in terms of continental drift. The ancestral ratite population existed at a time when the southern continents of South America, Africa, and Australia (and their major offshore islands) were joined as a single land mass called Gondwana. As the continents moved apart as a result of plate tectonics, the early ratite populations were carried with them. Subsequent speciation on each continent and some of the islands produced the variety of forms shown here. The 50 species of tinamou (see chart below) from South America, are considered a sister group to the ratites even though they can fly, because they possess the archaic palate. This relationship is confirmed by DNA sequence tests. The diagram below shows a possible phylogenetic tree based upon comparisons of mitochondrial DNA sequences. This view has been supported by the extensive comparison of skeletons from the different ratite species.



* Lambert *et al.* 2004. "Ancient DNA solves sex mystery of moa." Australasian Science, 25(8), Sept. 2004, pp. 14-16.

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1. (a) Describe three physical features distinguishing all ratities from most other birds: _____

_____

(b) What primitive feature is shared by ratites and tinamou? _____

2. Describe two anatomical changes, common to all ratites, which have evolved as a result of flightlessness. For each, describe the selection pressures for the anatomical change:

(a) Anatomical change: _____

Selection pressure: _____

_____

(b) Anatomical change: _____

Selection pressure: _____

_____

3. Which ancient super-continent did the ancestral ratite population inhabit? _____

4. (a) The extinct elephantbird from Madagascar is thought to be very closely related to another modern ratite. Based purely on the **geographical distribution** of ratites, Which modern species is the most likely relative?

_____

(b) Explain why you chose the modern ratite in your answer to (a) above: _____

_____

(c) Draw lines on the diagram at the bottom of the previous page to represent the divergence of the elephantbird from the modern ratite you have selected above.

5. (a) Name two other flightless birds that are not ratites: _____

(b) Why are these other flightless species not considered part of the ratite group? _____

_____

_____

6. Eleven species of moa is an unusually large number compared to the species diversity of kiwi, the other ratite group found in New Zealand. Moa are classified into at least four genera, whereas kiwi have only one genus. The diets of the moa and kiwi are thought to have had a major influence on each group's capacity to diverge into separate species and genera. Moa were herbivorous, whereas kiwi are nocturnal feeders, feeding on invertebrates in the leaf litter. Explain why, on the basis of their diet, moa diverged into many species, whereas kiwi diverged little:

_____

_____

_____

_____

7. The DNA evidence suggests that New Zealand had two separate invasions of ratites, an early invasion from moa (before the breakup of Gondwana) followed by a second invasion of the ancestors of the kiwi. Describe a possible sequence of events that could account for this:

_____

_____

_____

_____

8. The common ancestors of divergent groups are labelled (**A**-**L**) on the diagram at the bottom of the previous page. State the **letter** identifying the **common ancestor** for:

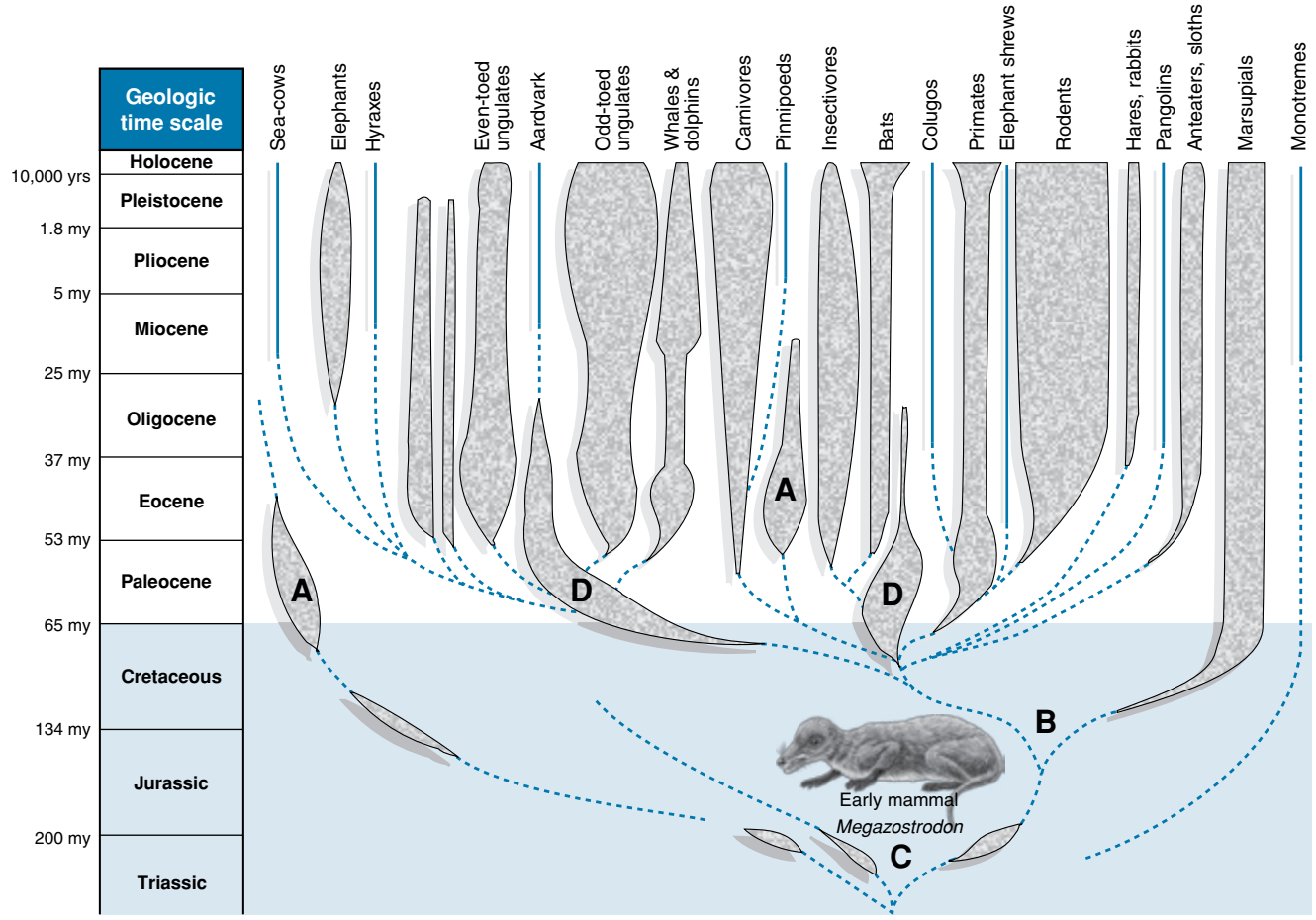
(a) Kiwi and the Australian ratites: _____ (b) Kiwi and moa: _____

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242 Adaptive Radiation in Mammals

Key Idea: An ancestral mammal group underwent adaptive radiation about 80 mya and diversified into all the mammalian groups seen today.

Adaptive radiation is diversification among the descendants of a single ancestral group to occupy different niches. Mammals underwent an extensive adaptive radiation following the extinction of the dinosaurs. Most of the modern mammalian groups became established very early on. The diagram below shows the divergence of the mammals into major orders, many occupying niches left vacant by the dinosaurs. The vertical extent of each grey shape shows the time span for which a particular order has existed. Those that reach the top of the chart have survived to the present day. The width of a grey shape shows how many species existed at any given time. The dotted lines indicate possible links between the orders for which there is no direct fossil evidence.



1. In general terms, discuss the **adaptive radiation** that occurred in mammals: _____

_____

_____

2. Name the term that you would use to describe the animal groups at point **C** (above): _____

3. Explain what occurred at point **B** (above): _____

_____

4. Describe two things that the animal orders labelled **D** (above) have in common:

   (a) _____

   (b) _____

5. Identify the two orders that appear to have been most successful in terms of the number of species produced:

_____

6. Explain what has happened to the mammal orders labelled **A** in the diagram above: _____

_____

7. Name the geological time period during which there was the most adaptive radiation: _____

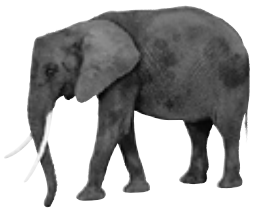
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8. What event was likely to have been instrumental in the rapid adaptive radiation of the mammals and why?

_____

9. There are 18 orders of placental mammals (or 17 in schemes that include the pinnipeds within the Carnivora). Their names and a brief description of the type of mammal belonging to each group is provided below. Identify and label each of the diagrams with the correct name of their Order:

Orders of Placental Mammals



1 _____



2 _____



3 _____



4 _____



5 _____



6 _____

Order	Description
Insectivora	Insect-eating mammals
Macroscelidae	Elephant shrews (formerly classified with insectivores)
Chiroptera	Bats
Cetacea	Whales and dolphins
Pholidota	Pangolins
Rodentia	Rodents
Probiscidea	Elephants
Sirenia	Sea-cows (manatees)
Artiodactyla	Even-toed hoofed mammals
Dermoptera	Colugos
Primates	Primates
Xenarthra	Anteaters, sloths, and armadillos
Lagomorpha	Pikas, hares, and rabbits
Carnivora	Flesh-eating mammals (canids, rac coons, bears, cats)
Pinnipedia	Seals, sealions, walruses. (Often now included as a sub-order of Carnivora).
Tubulidentata	Aardvark
Hyracoidea	Hyraxes
Perissodactyla	Odd-toed hoofed mammals



7 _____



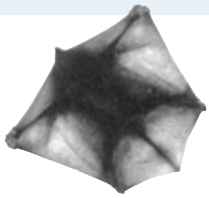
8 _____



9 _____



10 _____



11 _____



12 _____



13 _____



14 _____



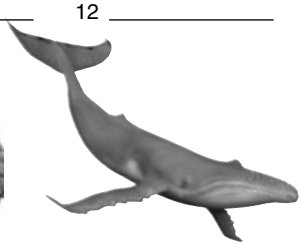
15 _____



16 _____



17 _____



18 _____

10. For each of three named **orders** of placental mammal, describe one **adaptive feature** that allows it to exploit a different niche from other placentals, and describe a **biological advantage** conferred by the adaptation:

(a) Order: _____ Adaptive feature: _____

Biological advantage: _____

_____

(b) Order: _____ Adaptive feature: _____

Biological advantage: _____

_____

(c) Order: _____ Adaptive feature: _____

Biological advantage: _____

_____

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243 Convergent Evolution

Key Idea: Evolution in response to similar selection pressures can result in unrelated species appearing very similar.

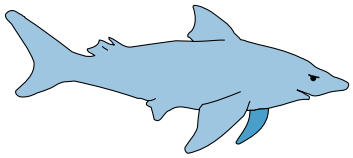
Convergent evolution (convergence) describes the process by which species from different evolutionary lineages come to resemble each other because they have similar ecological roles, and natural selection has shaped similar adaptations. It can be difficult to distinguish convergent and parallel evolution, as both produce similarity of form. Generally, similarity arising in closely related lineages (e.g. within marsupial mice) is regarded as parallelism, whereas similarity arising in more distantly related taxa is convergence (e.g. similarities between marsupial and placental mice).

Convergence in Swimming Form

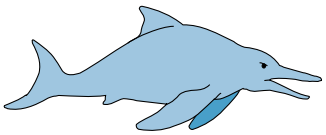
Not all similarities between species are the result of common ancestry. Selection pressures to solve similar problems in particular environments may result in similarity of form and function in unrelated (or distantly related) species. The development of succulent forms in unrelated plant groups (*Euphorbia* and the cactus family) is an example of **convergence** in plants. In the example (right), the selection pressures of the aquatic environment have produced a similar **streamlined** body shape in unrelated vertebrate groups. Icthyosaurs, penguins, and dolphins each evolved from terrestrial species that took up an aquatic lifestyle. Their general body form has evolved to become similar to that of the shark, which has always been aquatic. Note that flipper shape in mammals, birds, and reptiles is a result of convergence, but its origin from the pentadactyl limb is an example of **homology**.

Analogous Structures

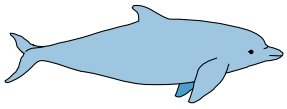
Analogous structures (or **homoplasies**) are those that have the same function and often the same appearance, but different origins. The example (right) shows the structure of the **eye** in two unrelated taxa (mammals and cephalopod molluscs). The eye appears similar, but has evolved independently. The **wings** of birds and insects are also analogous structures. The wings have the same function, but the two taxa do not share a common ancestor. *Longisquama*, a lizard-like creature that lived about 220 mya, also had 'wings' that probably allowed gliding between trees. These 'wings' were not a modification of the forearm (as in birds), but highly modified long scales or feathers extending from its back.



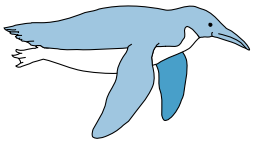
Fish: Shark



Reptile: Icthyosaur (extinct)



Mammal: Dolphin



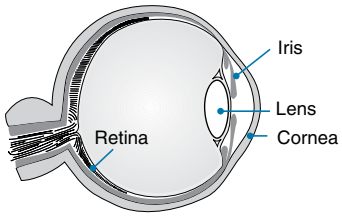
Bird: Penguin

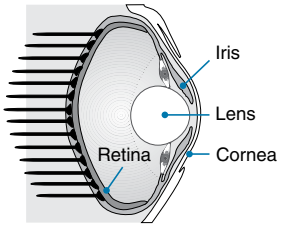


Mammalian eye



Octopus eye





1. In the example above illustrating convergence in swimming form, describe two ways in which the body form has evolved in response to the particular selection pressures of the aquatic environment:

   (a) _____

   _____

   (b) _____

   _____

2. Describe two of the selection pressures that have influenced the body form of the swimming animals above:

   (a) _____

   (b) _____

3. When early taxonomists encountered new species in the Pacific region and the Americas, they were keen to assign them to existing taxonomic families based on their apparent similarity to European species. In recent times, many of the new species have been found to be quite unrelated to the European families they were assigned to. Explain why the traditional approach did not reveal the true evolutionary relationships of the new species:

   _____

   _____

   _____

   _____

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4. For each of the paired examples, briefly describe the adaptations of body shape, diet and locomotion that appear to be similar in both forms, and the likely selection pressures that are acting on these mammals to produce similar body forms:



Convergence Between Marsupials and Placentals

Marsupial and **placental** mammals diverged very early in mammalian evolution (about 120 mya), probably in what is now the Americas. Marsupials were widespread throughout the ancient supercontinent of Gondwana as it began to break up through the Cretaceous, but then became isolated on the southern continents, while the placentals diversified in the Americas and elsewhere, displacing the marsupials in most habitats around the world. Australia's isolation from other landmasses in the Eocene meant that the Australian marsupials escaped competition with the placentals and diversified into a wide variety of forms, ecologically equivalent to the North American placental species.



Some older sources cite this example as one of parallelism, rather than convergence. However, a greater degree of morphological difference than now once separated the ancestors of the placental and marsupial lineages being compared, making this a case of convergence, not parallelism.

Marsupial Mammals		Placental Mammals
Wombat 	(a) Adaptations: Rodent-like teeth, eat roots and above ground plants, and can excavate burrows. Selection pressures: Diet requires chisel-like teeth for gnawing. The need to seek safety from predators on open grassland.	Woodchuck 
Flying phalanger 	(b) Adaptations: Selection pressures:	Flying squirrel 
Marsupial mole 	(c) Adaptations: Selection pressures:	Mole 
Marsupial mouse 	(d) Adaptations: Selection pressures:	Mouse 
Tasmanian wolf (tiger) 	(e) Adaptations: Selection pressures:	Wolf 
Long-eared bandicoot 	(f) Adaptations: Selection pressures:	Jack rabbit 

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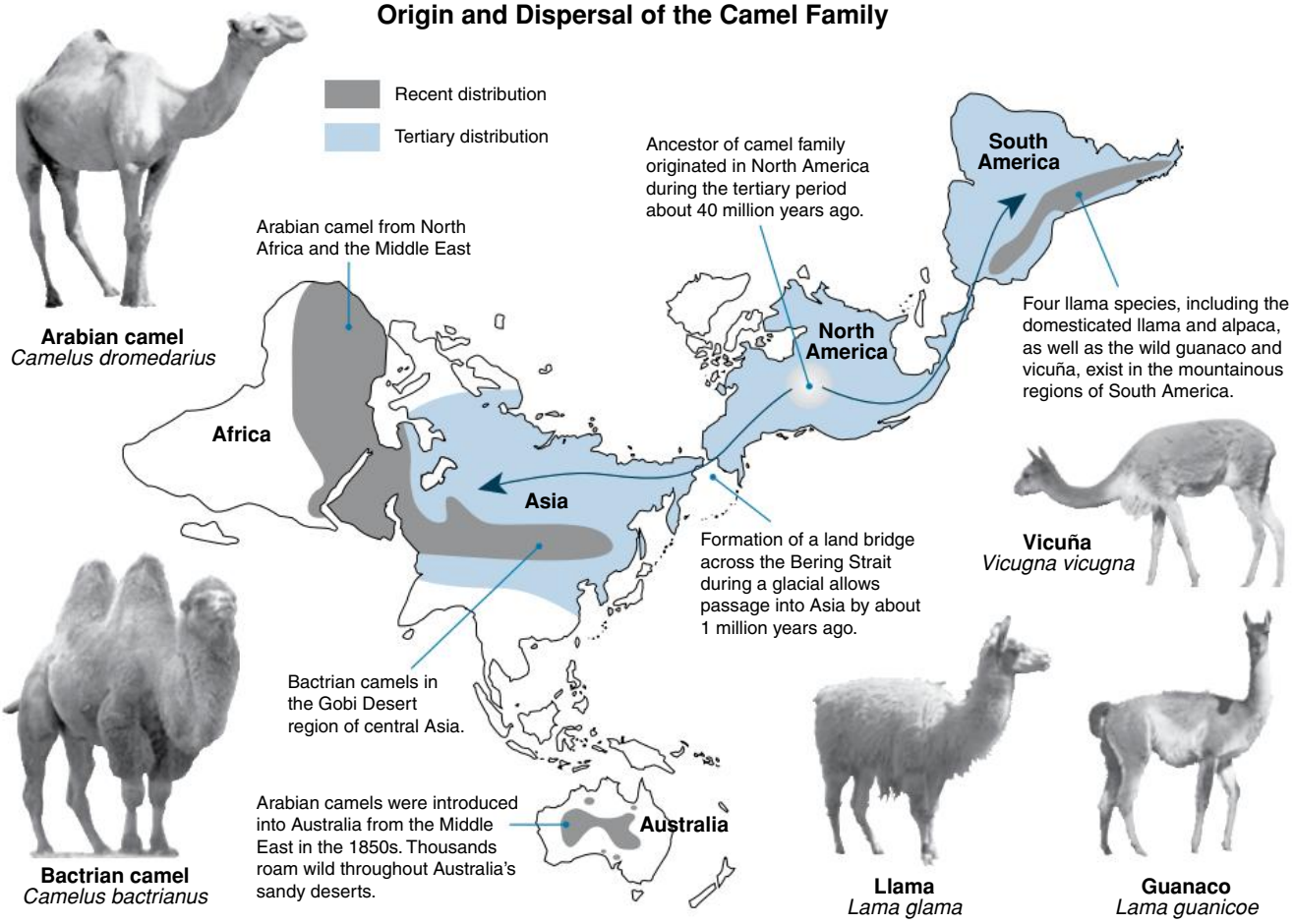
244 Speciation and Dispersal of the Camelidae

Key Idea: The current fragmented distribution of the camel family can be explained by natural phenomena such as migration, plate tectonics, and changes in sea level.

The camel family, Camelidae, consists of six modern-day species on three continents: Asia, Africa and South America. Geophysical phenomena such as plate tectonics and glacial cycles have determined their present, fragmented distribution. Three principles about the dispersal and distribution of land animals are:

- When very closely related animals were present at the same time in widely separated parts of the world, it is highly probable that there was no barrier to their movement in between the localities in the past.
- The most effective barrier to movement of land animals (especially mammals) was a sea between continents.
- The scattered distribution of living species can be explained by migration out of their original range or extinction in regions between the current populations.

Origin and Dispersal of the Camel Family



1. Arabian camels are found wild in the Australian outback.

   (a) How did they get there? _____

   (b) Why they were absent during prehistoric times? _____

2. The camel family originated in North America. Suggest why there are no members of the family in North America now:

   _____

3 Suggest how early camels managed to get to Asia from North America: _____

   _____

   _____

4. (a) Describe the present distribution of the camel family: _____

   _____

   (b) Explain why it is scattered (discontinuous): _____

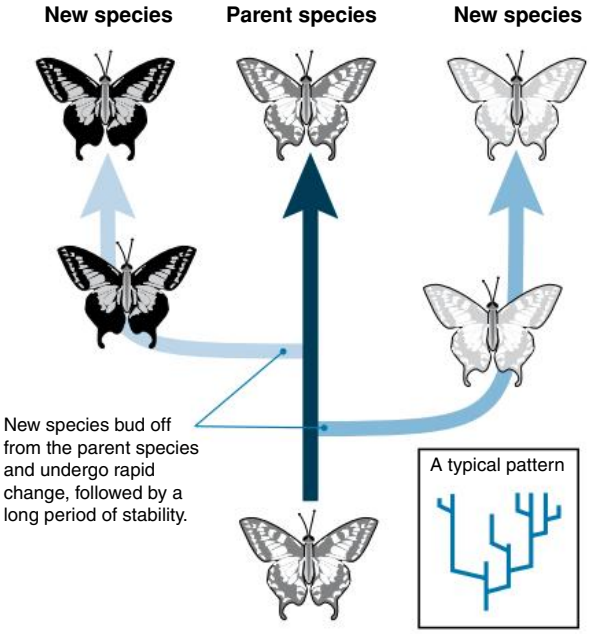
   _____

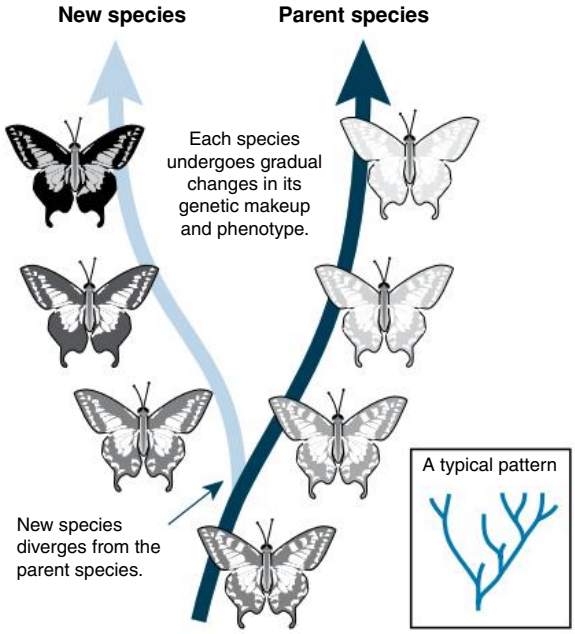
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245 The Rate of Evolutionary Change

Key Idea: A new species may form gradually over a long period of time, or relatively suddenly.

Two main models have been proposed for the rate at which evolution occurs: **gradualism** and **punctuated equilibrium**. It is likely that both mechanisms operate at different times and in different situations. Interpretations of the fossil record vary depending on the time scales involved. During its formative millennia, a species may have accumulated changes gradually (e.g. over 50,000 years). If that species survives for 5 million years, the evolution of its defining characteristics would have been compressed into just 1% of its evolutionary history. In the fossil record, the species would appear quite suddenly.





Punctuated Equilibrium

There is abundant evidence in the fossil record that, instead of gradual change, species stayed much the same for long periods of time (called stasis). These periods were punctuated by short bursts of evolution which produce new species quite rapidly. According to the punctuated equilibrium theory, most of a species' existence is spent in **stasis** and little time is spent in active evolutionary change. The stimulus for evolution occurs when some crucial aspect of the environment changes.

Phyletic Gradualism

Phyletic gradualism assumes that populations slowly diverge by accumulating adaptive characteristics in response to different selective pressures. If species evolve by gradualism, there should be transitional forms seen in the fossil record, as is seen with the evolution of the horse. Trilobites, an extinct marine arthropod, are another group of animals that have exhibited gradualism. In a study in 1987 a researcher found that they changed gradually over a three million year period.

1. What rate of environmental change would support the following paces of evolution?

   (a) Punctuated equilibrium: _____

   _____

   (b) Gradualism: _____

   _____

2. In the fossil record of early human evolution, species tend to appear suddenly, linger for often very extended periods before disappearing suddenly. There are few examples of smooth inter-gradations from one species to the next. Which of the above models best describes the rate of human evolution?

   _____

3. Some species show apparently little evolutionary change over long periods of time (hundreds of millions of years).

   (a) Name two examples of such species: _____

   (b) What term is given to this lack of evolutionary change? _____

   (c) Such species are often called 'living fossils'. Why might they have been described in this way? _____

   _____

   (d) Why would such species change so little over evolutionary time? _____

   _____

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246 Extinction

Key Idea: Extinction is a natural process. Mass extinctions involve the extinction of large numbers of species and genera in a geologically short period of time.

Most species that have ever lived are now extinct. Extinction is an important process in evolution as it provides opportunities, in the form of vacant niches, for new species. Background extinction is the steady rate of species turnover in a taxonomic group. Superimposed on this constant rate of background extinction are catastrophic events or **mass extinctions** that wipe out vast numbers of species in geologically brief periods of time. The diagram below shows how the number of species has varied over the history of life on Earth. The number of species is indicated on the graph by families: a taxonomic group comprising many genera and species. There have been five major extinction events and a sixth event, which began in the Late Pleistocene and continues today.

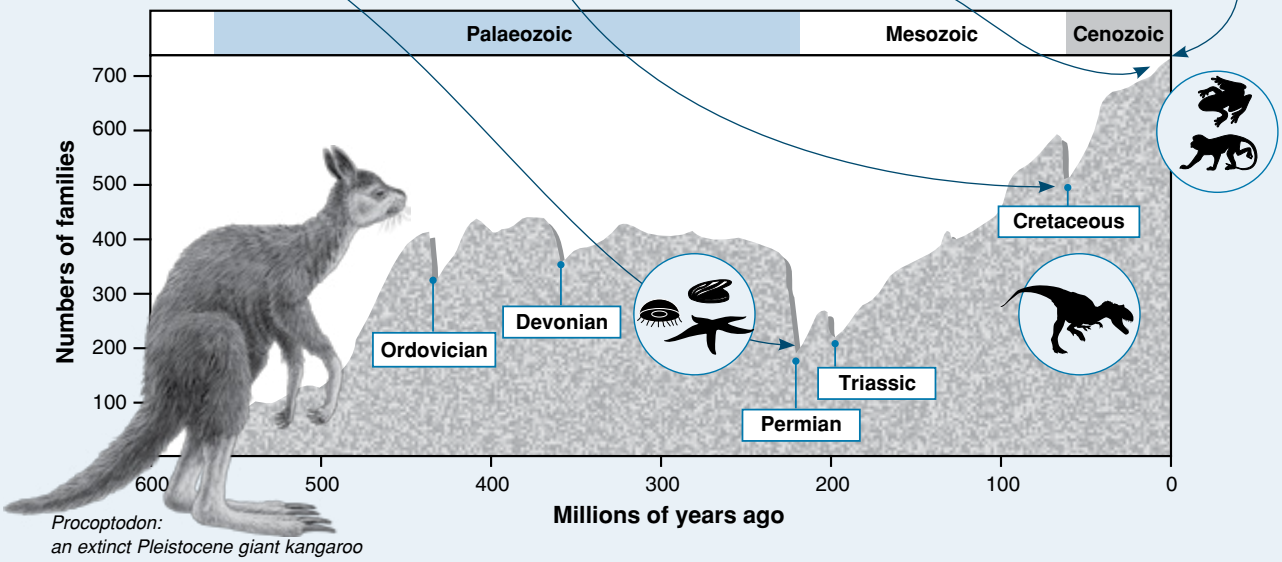
Major Mass Extinctions

The Permian extinction
(250 million years ago)
This was the most devastating mass extinction of all. Nearly all life on Earth perished, with 90% of marine species and probably many terrestrial ones also, disappearing from the fossil record. This extinction event marks the **Paleozoic-Mesozoic** boundary.

The Cretaceous extinction
(65 million years ago)
This extinction event marks the boundary between the Mesozoic and Cenozoic eras. More than half the marine species and many families of terrestrial plants and animals became extinct, including nearly all the dinosaur species (the birds are now known to be direct descendants of the dinosaurs).

Quaternary extinction
(15,000-10,000 years ago)
This first phase of the sixth extinction is marked by the extinction of many species of megafauna (giant mammal), although there were few extinctions of entire families. It is known as the **Pleistocene overkill** because it was associated with the hunting activities of prehistoric humans. Many large marsupials in Australia and placental species elsewhere became extinct.

The sixth extinction
(second phase occurring now)
The current phase of the sixth extinction is largely due to human destruction of habitats (e.g. coral reefs, tropical forests) and pollution. It is considered far more serious and damaging than some earlier mass extinctions because of the speed at which it is occurring. The increasing human impact is making biotic recovery difficult.



Procoptodon:
an extinct Pleistocene giant kangaroo

1. Describe the main features (scale and type of organisms killed off) of each of the following major extinction events:

   (a) Permian extinction: _____

   _____

   (b) Cretaceous extinction: _____

   _____

   (c) Megafaunal extinction: _____

   _____

2. How has human activity contributed to the most recent mass extinction? _____

   _____

3. In general terms, describe the effect of past mass extinctions on the evolution of surviving species: _____

   _____

   _____

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247 KEY TERMS: Mix and Match

INSTRUCTIONS: Test your vocabulary by matching each term to its correct definition, as identified by its preceding letter code. This activity tests your knowledge of the vocabulary used in this and the previous chapter.

adaptive radiation .....

allopatric .....

analogous structure .....

chronometric dating .....

common ancestor .....

convergent evolution .....

divergent evolution .....

extinction .....

fossil .....

hybrid .....

macroevolution .....

mass extinction .....

microevolution .....

natural selection .....

parallel evolution .....

phyletic gradualism .....

phylogeny .....

postzygotic isolating mechanism .....

prezygotic isolating mechanism .....

punctuated equilibrium .....

relative dating .....

reproductive isolation .....

speciation .....

species .....

sympatric .....

A A feature of different species that are similar in function but not necessarily in structure and in which the similarity is not the result of common ancestry.

B An offspring resulting from the mating of two genetically dissimilar parents, e.g. two inbred lines or even two different species.

C Speciation in which the populations are physically separated. A term for physically separated populations.

D Evolutionary process in which a species or related species follow different evolutionary pathways to eventually become less related.

E Changes in the allele frequencies of a species gene pool through natural selection, gene flow, mutation, and genetic drift.

F The independent evolution of similar traits, starting from a similar ancestral state.

G The most recent individual from which all organisms in the group are directly descended.

H The process by which traits become more or less common in a population due to the differential survival or reproduction of their bearers.

I Determining an approximate age for an archaeological or palaeontological site or artefact based on its physical or chemical properties of the materials.

J A reproductive isolating mechanism that occurs before the formation of a zygote.

K Evolution in unrelated species occupying similar niches that causes them to arrive at similar structural, physiological and behavioural solutions.

L The preserved remains or traces of organisms from the remote past.

M Group or population of individuals that can interbreed to produce viable offspring.

N Speciation as a result of reproductive isolation without any physical separation of the populations, i.e. populations remain within the same range. A term for populations sharing the same range.

O The evolutionary history or genealogy of a group of organisms. Often represented as a 'tree' showing descent of new species from the ancestral one.

P Large evolutionary patterns above the species level, e.g. adaptive radiations.

Q A model for the evolution of lineages in which long periods of stasis are interrupted by brief periods of rapid speciation.

R The complete dying out of a species so that there are no representatives of the species remaining anywhere.

S An episode of large-scale extinction affecting many different groups of organisms within a short interval of geological time.

T The situation in which members of a group of organisms breed with each other but not with members of other groups.

U A reproductive isolating mechanism that occurs after the formation of the zygote.

V A model for the evolution of different forms over a long period of time but with only slight changes occurring between successive generations. Also called anagenesis.

W The division of one species, during evolution, into two or more separate species.

X The science determining the relative order of past events, without necessarily determining their absolute age.

Y A form of divergent evolution in which there is rapid speciation of one ancestral species to fill many different ecological niches.

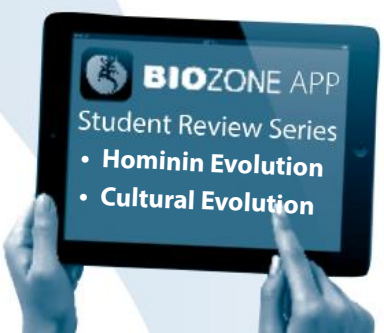
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QLD As required
VCE Unit 4 (2)
WA ATAR Unit 3
SA N/A

Human Evolution

Key terms

ape
Au. afarensis
Acheulian (tool)
Ardipithecus ramidus
Australopithecus spp.
bipedal (bipedalism)
Broca's area
carrying angle
Dmanisi fossil
gracile
hominid
hominoid
Homo
H. erectus
H. ergaster
H. floresiensis
H. habilis
H. heidelbergensis
H. neanderthalensis
H. sapiens
Mesolithic
multiregional hypothesis
Neolithic
Orrorin tugenensis
Out of Africa hypothesis
primate
Palaeolithic
Paranthropus spp.
prehensile
prognathic
replacement hypothesis
robust
sexual dimorphism
valgus angle
Wernicke's area



Our primate heritage

Learning aims and skills

Activity number

- ☐ 1 Appreciate that the classification of primates has several revisions in recent decades to accommodate new genetic evidence. Recognise strepsirrhines, tarsiers, New World monkeys, Old World monkeys, hominoids, hominids. — 248
- ☐ 2 Describe the physical and behavioural characteristics of primates, including features of the primate hand. Distinguish between primate groups in therms of their physical and behavioural characteristics. — 249 250





The physical evolution of humans

Learning aims and skills

Activity number

- ☐ 3 State the full classification of modern humans. Describe the anatomical and behavioural features that are unique to humans. — 248 252
- ☐ 4 Explain how relative and chronometric (absolute) dating methods are used in establishing a timeline for human evolution — 251
- ☐ 5 Discuss the hypotheses for the origin and dispersal of modern humans and the evidence for these. — 253 256
- ☐ 6 Describe the selection pressures on early hominins and the benefits of reducing body hair and adopting bipedalism as a form of locomotion. — 254 260
- ☐ 7 Describe the anatomical features associated with bipedalism, including the significance of the valgus (carrying) angle. — 261
- ☐ 8 Describe the anatomical features, geographical distribution, evolutionary relationships, and possible niche differences of the earliest hominins: *Ardipithecus*, *Orrorin*, *Australopithecus* spp., *Paranthropus* spp. — 255-259
- ☐ 9 Relate the evolution of the australopithecines, and later *Homo*, to changes in habitat and a shift in the resources they exploited. — 254 259 260
- ☐ 10 Describe the distinguishing characteristics unique to genus *Homo*. Discuss the biological and cultural evolution of species in the genus *Homo*. Compare and contrast these hominins with respect to skeletal structure, cranial capacity, fossil ages and regional locations, and inferred culture. — 255 262 263 264

The cultural evolution of humans

Learning aims and skills

Activity number

- ☐ 11 Describe trends and consequences of brain development, including increasing dependence on cooperation, art, spirituality, and language. — 263 264 269
- ☐ 12 Describe the features of the various Palaeolithic tool cultures, identifying the advancements in tool technology made at each stage. — 265 266
- ☐ 13 Discuss how the controlled use of fire and development of permanent settlements influenced and benefited human cultural evolution. — 267 268
- ☐ 14 Discuss cultural evolution in the Mesolithic and Neolithic ages. — 270 271

248 Primate Classification

Key Idea: Humans are primates, with genetic evidence showing chimpanzees to be our closest living relatives.

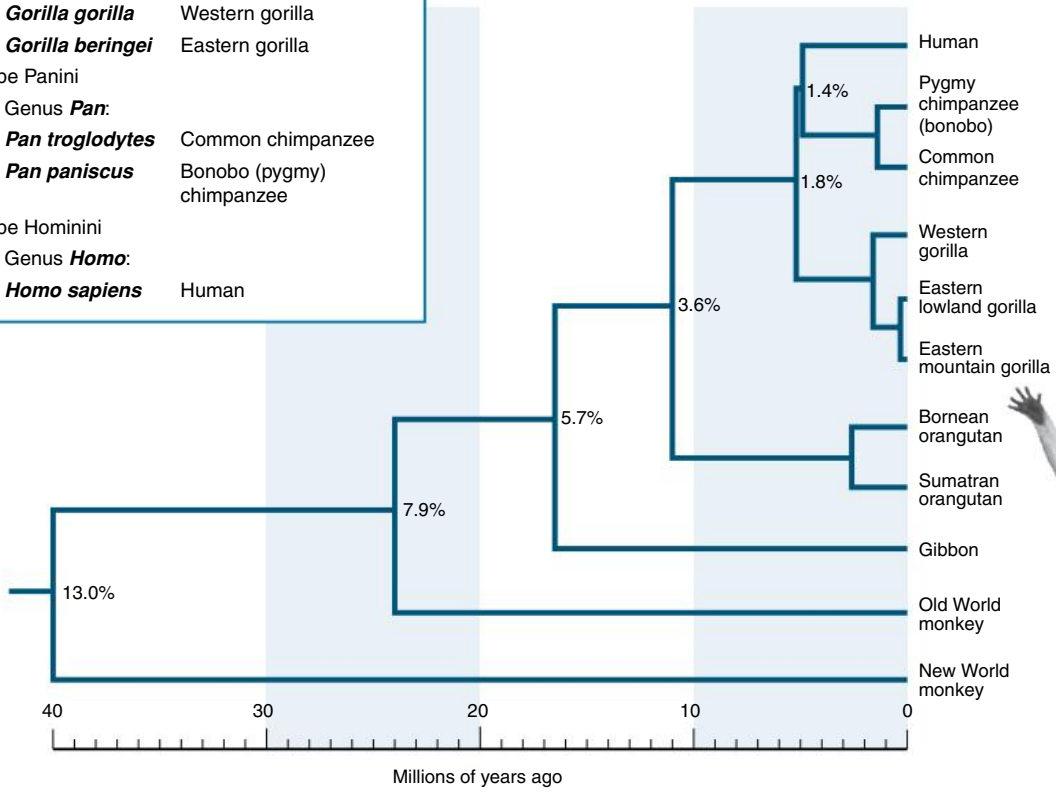
The classification of primates has been considerably revised several times in the last few decades. Classification should reflect evolutionary history, and the modern classifications of primates are supported by genetic, as well as morphological, evidence of phylogeny. Current classifications favour two primate suborders: the Strepsirrhini (lemurs and lorises) and the Haplorhini (tarsiers, monkeys, apes, and humans). Within the haplorhines, the classification of the apes has been particularly controversial, and heavily influenced by the historical perception that humans were special and worthy of their own family. Based on genetic evidence, chimps and gorillas are more closely related to humans than to orangutans, and chimps are more closely related to humans than they are to gorillas.

Superfamily Hominoidea

Family Hylobatidae:	{various gibbon species}
Family Hominidae:	
Subfamily Ponginae	
Genus ***Pongo***:	
Pongo pygmaeus	Bornean orangutan
Pongo abelii	Sumatran orangutan
Subfamily Homininae	
Tribe Gorillini	
Genus ***Gorilla***:	
Gorilla gorilla	Western gorilla
Gorilla beringei	Eastern gorilla
Tribe Panini	
Genus ***Pan***:	
Pan troglodytes	Common chimpanzee
Pan paniscus	Bonobo (pygmy) chimpanzee
Tribe Hominini	
Genus ***Homo***:	
Homo sapiens	Human

Primate family tree

The diagram below presents a classification of hominoids (apes and humans) based on genetic differences. The percentages next to each of the points where a split occurs indicates the amount of difference in the total genetic makeup (genomes) of the two groups being considered, e.g the genome of the gibbons compared to the rest of the apes (orangutans, gorillas, chimpanzees) and humans differs by 5.7%. A large genetic difference between any two groups implies that they are distantly related, whereas small genetic differences suggest they share a recent common ancestor.







1. According to the diagram above, which shows relatedness according to genetic similarity:

   (a) Which hominoid group is most closely related to the two chimpanzee species? _____

   (b) Name the two chimpanzee species: _____

2. From the diagram, determine how long ago:

   (a) The two species of chimpanzee split from a common ancestor: _____

   (b) The chimpanzees split from the line to humans: _____

   (c) The African apes (and humans) split from the Asian apes (orangutans and gibbons): _____

3. Circle on the diagram the point where the African (Old World) monkeys split from the American (New World) monkeys:

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249 General Primate Characteristics

Key Idea: Primates exhibit unique, but quite generalised, morphological, physiological, and behavioural features.
Morphological features include five digits on the hands and feet, physiological features include a longer gestation than other mammals, and behavioural features include prolonged infant dependency and keen social behaviour.

The **gestation** (pregnancy period) in primates is longer than most other mammals. Primates typically have one young per pregnancy (below). Infancy is prolonged with longer periods of infant dependency and a large **parental investment** in each offspring. This nurturing increases the survival rate of the young and allows cultural development.



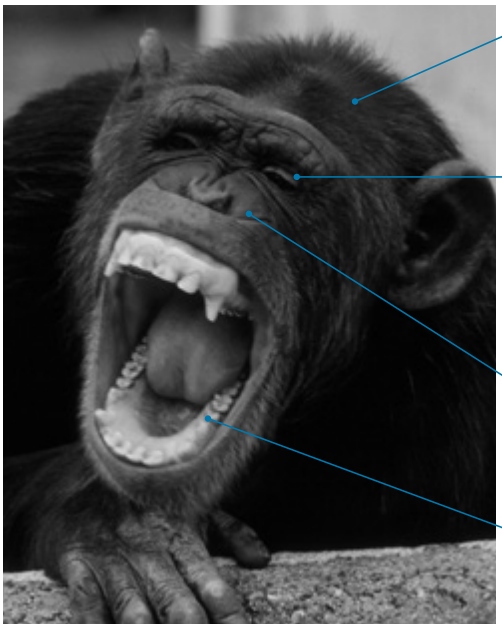




Primates generally live longer relative to most other mammals and there is a greater dependency on highly flexible learned behaviour. Primates tend to be **highly sociable** (above). Unusually for mammals, adult males of many primate species often associate permanently with the group.



Chimpanzees and **gorillas** spend more time out of trees than do either of the Asian apes. The chimpanzee above shows typical **knuckle-walking** behaviour. The relatively long arms facilitate this mode of locomotion.



The **brain** is large and generally more complex than in other mammals.

Vision is important, the visual areas of the brain are enhanced. Well developed binocular, stereoscopic vision provides overlapping visual fields and good depth perception. Colour vision is probably present in all primates, except specialised nocturnal forms.

A trend towards a **reduced snout** and flattened face and reduced olfactory regions in the brain. Baboons go against this trend, with a secondary increase in muzzle length.

Primates have a generalised **dental pattern** particularly in the back teeth. Unspecialised teeth enabled primates to adopt a flexible omnivorous diet.



Primates have a tendency toward **erectness**, particularly in the upper body, as seen in the gorilla, above. This tendency is associated with sitting, standing, leaping, and (in some) walking.



Gibbons are the smallest of the apes and are specialised to use **brachiation** (a technique of under-branch swinging), in combination with rapid climbing, midair leaps, and bipedal running, to move quickly through the forest.



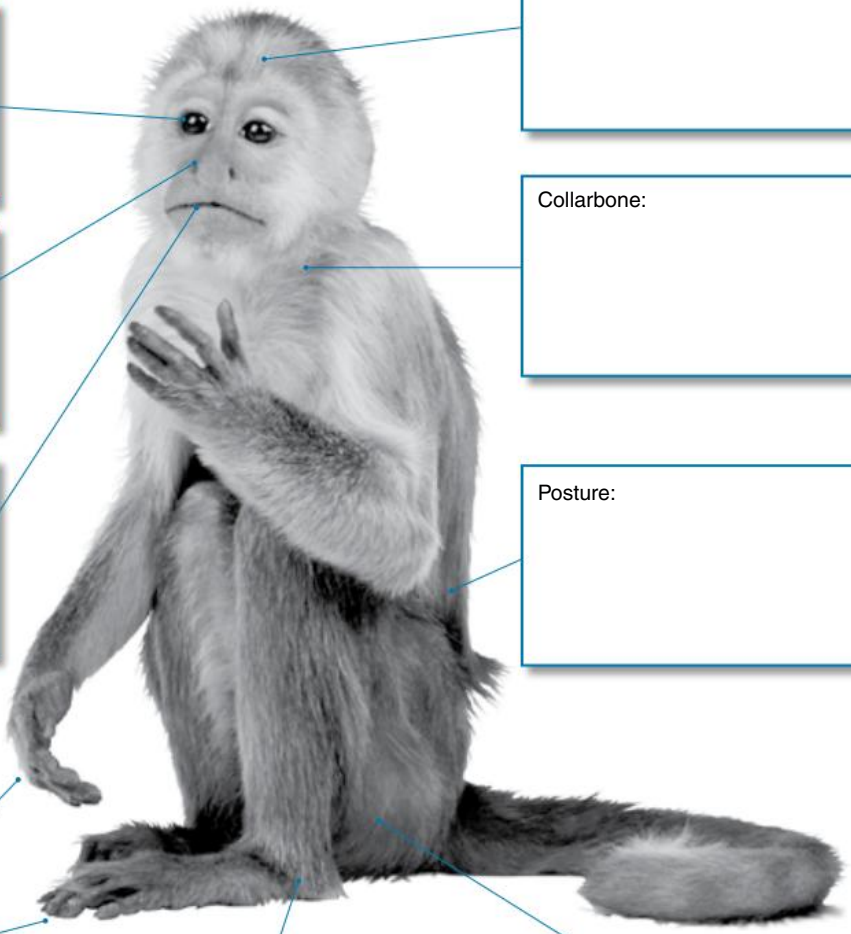
Monkeys walk **quadrupedally** on the palms of their hands and the soles of their feet. In the trees, they walk along the tops of branches, gripping them with their hands and feet.

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LINK 250 WEB 249 KNOW

1. On the diagram of the capuchin below, briefly describe the general physical characteristics of all primates as indicated:

The primate pictured is a **white-fronted capuchin monkey** (*Cebus albifrons*) from northern South America. These monkeys inhabit the mid-canopy deciduous, gallery forests.



Vision:

Face shape and snout:

Teeth shape and dental arrangement:

Brain size and specialisation:

Collarbone:

Posture:

Hands and feet:

Limb joints:

Reproduction:

Social organisation:

2. Humans belong to the order Primates. Describe the characteristics of humans that characterise their primate heritage:

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250 The Primate Hand

Key Idea: Primates have grasping hands, but the human hand is particularly developed with respect to dexterity and the manipulation of objects.

Primates have a grasping hand. They are able to pick things up, hold, and manipulate them, although the degree to which a primate can do this depends on the species. Humans have a highly advanced ability to manipulate objects with their hands because the thumb is very long relative to the hand.



Index finger and little finger are able to pivot and move towards each other, allowing the fingers to form around small objects.

The fingers have end tactile pads that contain huge numbers of nerve endings, producing a highly sensitive surface.

Large muscles, especially around the thumb, produce a powerful grip.

Nails are found on at least some digits in all modern primates.

Flexible joints in the hand allow it to flex, increasing dexterity.

Highly mobile thumb, able to touch all other fingers on the hand.

LBS



Chimpanzees are very capable tool users, but their dexterity is limited by the length of their hand compared to the thumb and the rigid wrist bones that limit wrist rotation.

New research suggests manipulating objects may not have been the only important factor in the evolution of the human hand. The human hand shape is one of the only configurations possible that maintains dexterity while allowing the hand to form a fist. Although a punch with a closed fist produces the same force as a slap with an open palm, a punch delivers the force to a smaller area, producing a much greater impact and potential for damage to an opponent.

Grips of the Human Hand



Precision/ power grip



Power grip



Power grip



Precision grip

All photos LBS

1. Name two possible selective pressures acting on the human hand: _____

_____

2. Explain why being able to manipulate objects is an evolutionary advantage: _____

_____

3. Identify two features that make the human hand so dexterous and compare them to a chimpanzee hand:

_____

_____

_____

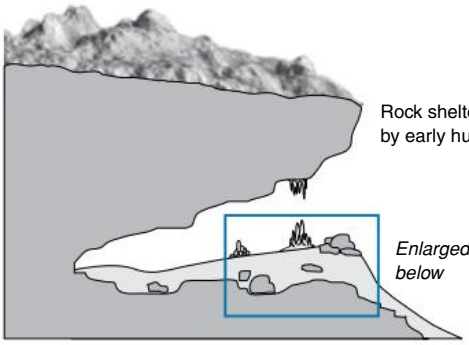
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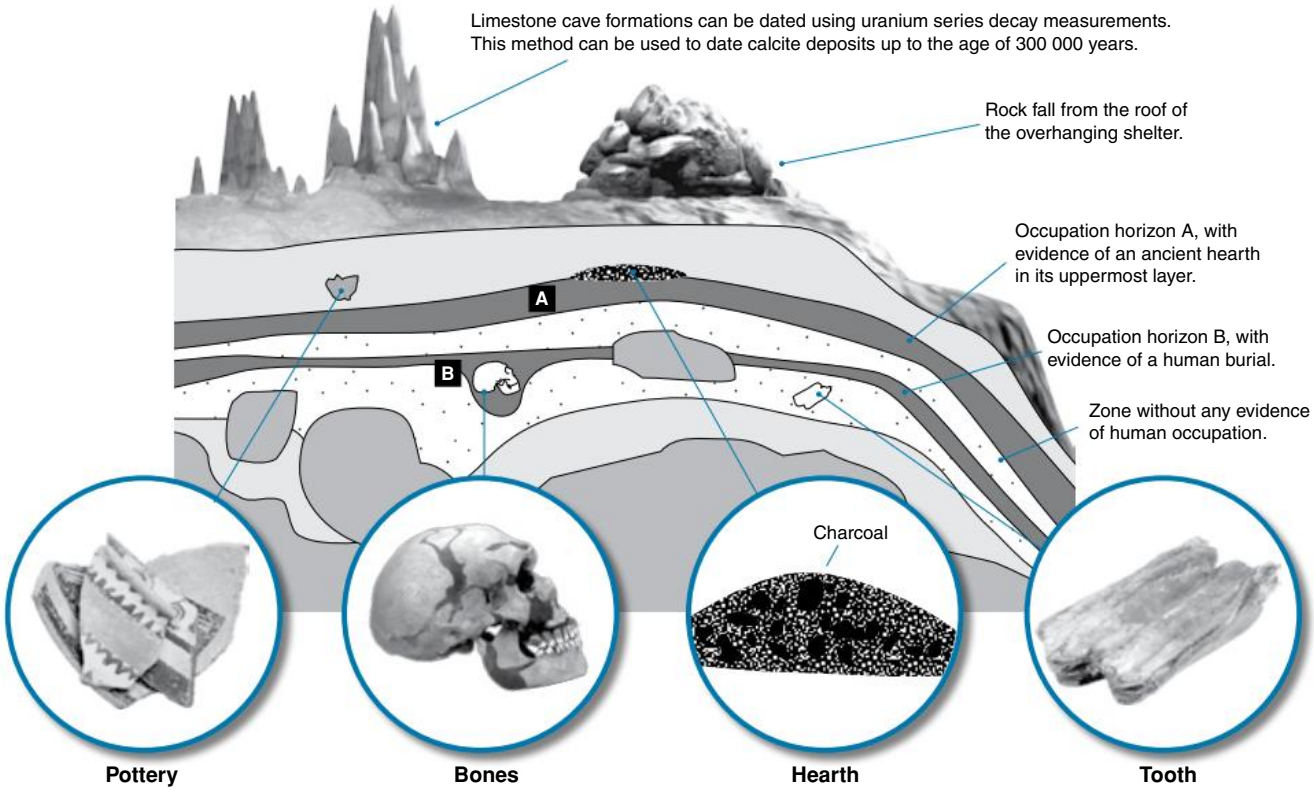
251 Dating a Fossil Site

Key Idea: A wide array of techniques can be used for dating artefacts, some of which show a high degree of reliability. The use of several appropriate techniques to date material improves the reliability of the date determined.

The diagram below shows a rock shelter typical of those found in the Dordogne Valley of Southwest France. Such shelters have yielded a rich source of Neanderthal and modern human remains. It illustrates the way human activity is revealed at archaeological excavations. Occupation sites included shallow caves or rocky overhangs of limestone. The floors of these caves accumulated the debris of natural rockfalls, together with the detritus of human occupation at various layers, called **occupation horizons**. Accurately dating these finds is important to understanding their significance.



Dating method	Dating range (years ago)	Datable materials
Radiocarbon (^{14}C)	1000 - 50,000+	Bone, shell, charcoal
Potassium-argon (K/Ar)	10,000 - 100 million	Volcanic rocks and minerals
Uranium series decay	less than 1 million	Marine carbonate, coral, shell
Thermoluminescence	less than 200 000	Ceramics (burnt clay)
Fission track	1000 - 100 million	Volcanic rock, glass, pottery
Electron spin resonance	2000 - 500,000	Bone, teeth, loess, burnt flint



Pottery
Pottery bowl dated at 7000 ± 350 years old.

Bones
Skull of an early human but unable to directly determine its age.

Hearth
The remains of an ancient fireplace was dated at 18 500 ± 1000 years old.

Tooth
A bison's tooth was dated at 45 000 ± 2500 years old.

1. Discuss the significance of **occupation horizons**: _____

_____

_____

_____

2. Determine the approximate date range for the items below: (Hint: take into account layers/artefacts with known dates)

   (a) The skull at point B: _____

   (b) Occupation horizon A: _____

3. Name the dating methods that could have been used to date each of the following, at the site above:

   (a) Pottery bowl: _____ (c) Hearth: _____

   (b) Skull: _____ (d) Tooth: _____

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Interpreting Fossil Sites

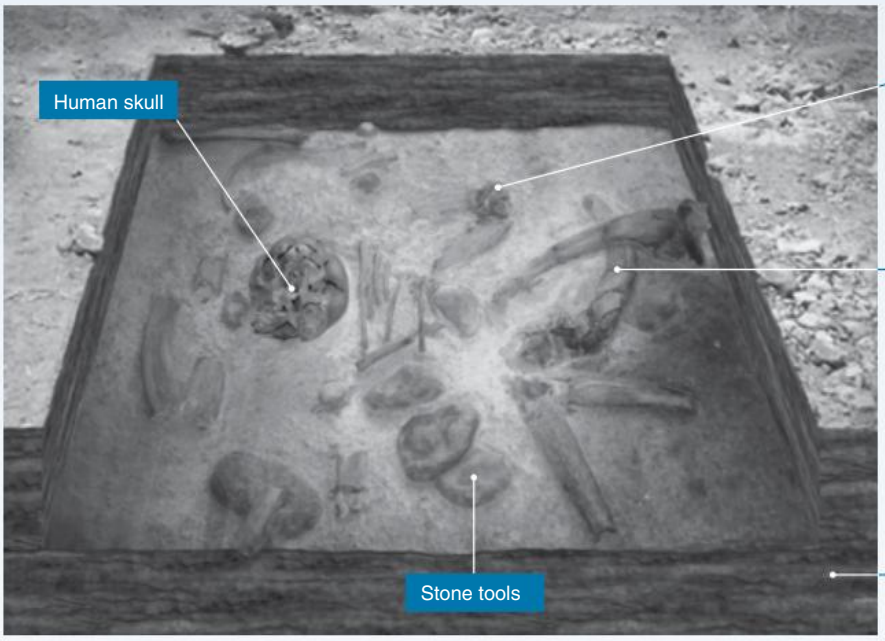


Photo: RA

Charcoal fragments (possible evidence of fire use and excellent for radiocarbon dating).

Bones from a large mammal with evidence of butchering (cut and scrape marks from stone tools). These provide information on the past ecology and environment of the hominins in question.

Excavation through rock strata (layers). The individual layers can be dated using both chronometric (absolute) and relative dating methods.



Searching for ancient human remains, including the evidence of culture, is the work of **paleoanthropologists**. Organic materials, such as bones and teeth, are examined and analyzed by physical anthropologists, while cultural materials, such as tools, weapons, shelters, and artworks, are examined by archaeologists. Both these disciplines, **paleoanthropology** and **archaeology**, are closely associated with other scientific disciplines, including **geochemistry** (for **chronometric dates**), **geology** (for reconstructions of past physical landscapes), and **paleontology** (for knowledge of the past species assemblages).

The reconstruction of a **dig site**, pictured above, illustrates some of the features that may be present at a site of hominin activity. Naturally, the type of information recovered from a site will depend on several factors, including the original nature of the site and its contents, the past and recent site environment, and earlier disturbance by people or animals. During its period of occupation, a site represents an interplay between additive and subtractive processes; building vs destruction, growth vs decay. Organic matter decays, and other features of the site, such as tools, can be disarranged, weathered, or broken down. The archaeologists goal is to maximise the recovery of information, and recent trends have been to excavate and process artefacts immediately, and sometimes to leave part of the site intact so that future work, perhaps involving better methodologies, is still possible.

4. Explain why paleoanthropologists date and interpret all of the remains at a particular site of interest (e.g. animal bones, pollen, and vegetation, as well as hominin remains):

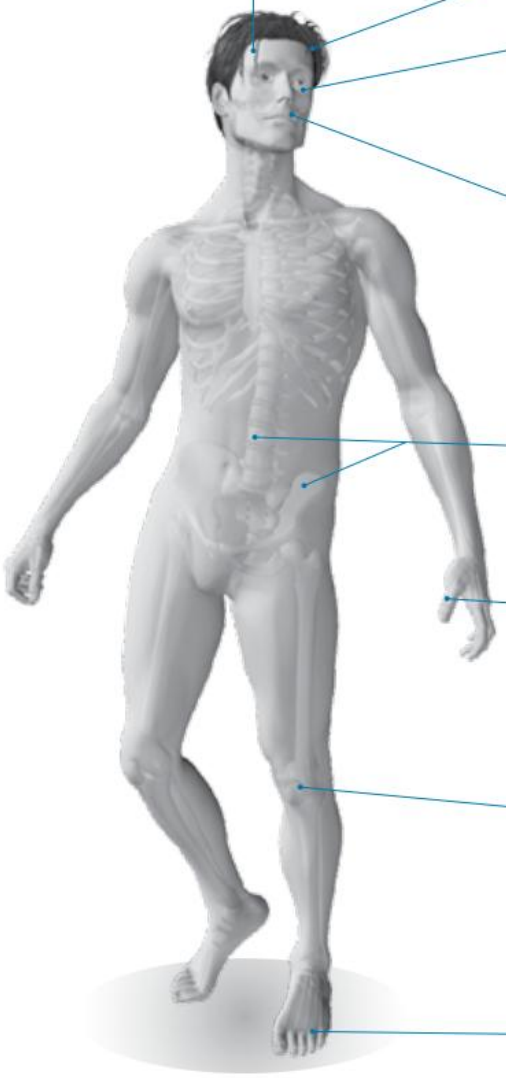
5. Discuss the importance of involving several scientific disciplines when interpreting a site of hominin activity:

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252 Recognising a Human Fossil

Key Idea: Fragmentary fossils make the identification of ancient humans and their ancestors difficult. Knowing the features that characterise a human is necessary for positive identification.

Humans have features that set them apart from other primates. Looking at the differences between modern humans and apes helps to identify trends in the evolutionary changes observed in the fossils of human ancestors.



Brain size and organisation: Enlarged, particularly in the frontal and occipital regions, associated with increased reliance on vision, advanced thinking (reasoning), and the production and interpretation of speech.

Skull shape: Articulated at the base rather than the rear, balanced upright on the spine. Brain case is rounded with reduced neck muscles.

Facial features: High forehead and characteristic upright face. Jaw has definite chin. Jaw is light and short with very reduced jaw muscles, hence there is no snout or sagittal crest.

Teeth size and shape: Small with reduced enamel thickness indicating a soft diet. Canines reduced, as they are no longer used as weapons. The cusps of the teeth (the bumps on the tooth surface) distinguish the type of tooth. Humans have five cusps on the molars of the lower jaw, but four on the molars of the upper jaw.

Spine and pelvis shape (male and female): S-shaped spine moves centre of gravity over the hips. Skull balanced on top of the spine.

Hands (degree of prehension, dexterity): Long, fully opposable thumb for precision grip.

Leg shape, hip joint (carrying angle) and knee joint: Bipedal gait with legs longer than the arms. Strong sets of walking muscles with short wide pelvis for muscle attachment. Buttress at knee joint to prevent sideways deflection of thigh muscles when walking. Leg bones thickened for increased load carrying. Head of femur and shaft angled to position knees along centre of gravity.

Feet adaptations to bipedalism: Arch with weight-bearing heel and ball. Big toe is forward thrusting and non-opposable.

1. Using the descriptions above, and information you have learned in this chapter, carry out the activities below:

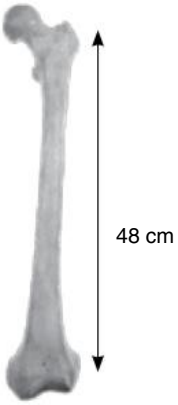
(a) Decide whether or not the jaw below is that of a modern human. Circle and annotate the photo to describe features to support your argument.



(b) The image below is of the top and rear of a non-human hominin skull. Annotate the photo to show the features that identify it as non-human.



(c) Annotate the photo to show the features that identify this as a human femur.



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253 Analysing Ancient DNA

Key Idea: Analysis of ancient human DNA can provide insight into the evolutionary relationships of human lineages.

Hominin fossil finds are rarely extensive enough to enable researchers to accurately classify or assess the morphology of the remains. In some instances, DNA analysis can be used to classify the specimens. New technologies have allowed ancient DNA to be extracted, enabling researchers to compare the genetic differences between species. The information from these comparative analyses can lead to new knowledge about human evolution and species relatedness.



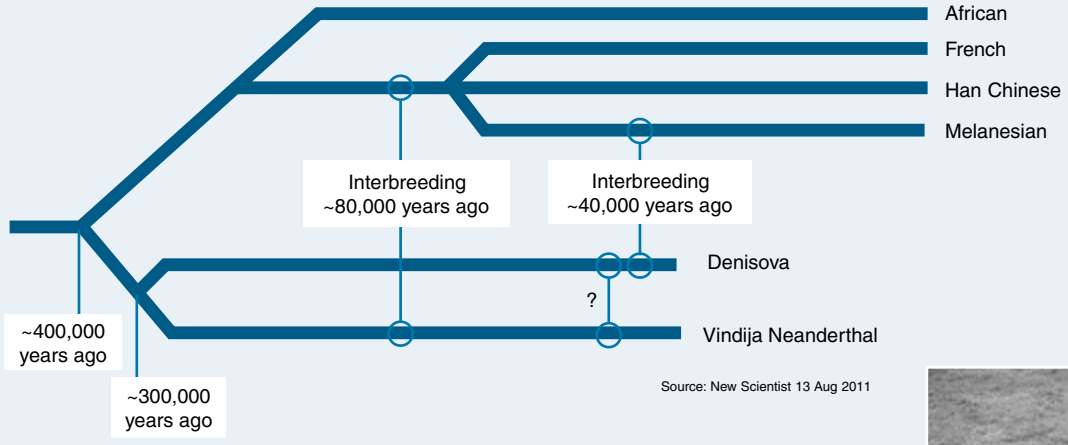
Obersachse

The Denisova cave, in the Altai mountains, Siberia, Russia

The Denisova Cave Finds

- In 2008, archeologists discovered a fragment of finger bone in the **Denisova cave**, in Siberia. The bone fragment belonged to a juvenile female (named X-woman).
- Artefacts, such as a bracelet, were found at the same level as the finger bone.
- In 2010, a molar tooth was found at a different level to the finger bone, indicating it belonged to a different individual. A toe bone found in 2011 was at the same level as the tooth.
- The molar found in the Denisova cave has unique characteristics, which are not present in the molars of Neanderthals or modern humans.
- Carbon dating estimates the age of the artefacts and bone fragment at 40,000 years.
- Fossil DNA degrades quite rapidly with increasing temperature and in acidic soil conditions. The cool temperatures within the Denisova cave preserved the DNA in the fossil fragments. The fossils contained very low levels of DNA contamination from other organisms.

Using Genome Analysis to Classify the Denisova Cave Fossils



Source: New Scientist 13 Aug 2011

Nuclear DNA analysis suggests the Denisova fossils belong to a previously unknown hominin species that existed at the same time as modern humans and Neanderthals, but was genetically distinct from them (above). The fossils are called the **Denisovans**, because they have not yet been formally classified.

Nuclear genome analysis suggests the Denisovans were a sister group to the Neanderthals. They probably shared a more recent common ancestor with Neanderthals (~300,000 years ago) than with present day humans (~400,000 years ago).

The Denisovan's interbred with the ancestors of the present day Melanesian's (right), and possibly with the Neanderthals, but not the ancestors of other present day populations, such as the Han Chinese. Melanesian DNA includes between 4% and 6% Denisovan DNA.



A Melanesian women

1. What are some of the limitations of relying solely on the **appearance** of fossils to classify new fossil finds?

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The Neanderthal Genome Project

Neanderthals appeared about 400,000 years ago, and disappeared 25,000-30,000 years ago. They lived in Europe and parts of western and central Asia. Neanderthals are the closest relative to modern humans, so there is considerable interest in mapping the Neanderthal genome. By comparing the Neanderthal genome to the genome of present-day humans, it may be possible to identify genes in modern humans that have been influenced by positive selection.

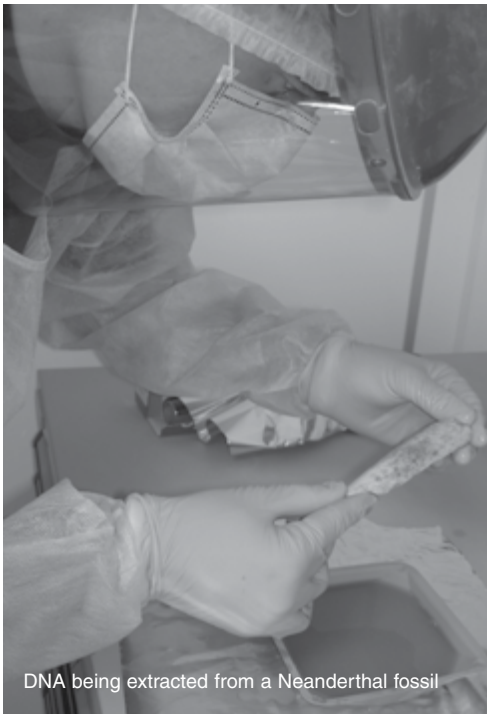


Front view Neanderthal Skull

Image: Bone clones



La Ferrassie Neanderthal Skull, France



DNA being extracted from a Neanderthal fossil

NIH

Difficulties in Analysing Neanderthal DNA

- The DNA is often degraded to small fragments less than 200 base pairs long. This makes it difficult to obtain sequence overlaps (critical for assembly of the genome).
- The DNA is often of poor quality because it has been chemically modified and degraded by the environment.
- Samples are often contaminated with the DNA of other organisms. Between 95-99% of the DNA obtained from the Neanderthal fossils analysed was from microbes that colonised the bone after the Neanderthal died. Great care must be taken by the researchers not to contaminate the sample with their own DNA.

The Findings

- The Neanderthal genome is about four billion base pairs long.
- The genome of modern human and Neanderthals is 99.7% identical.
- Interbreeding with anatomically modern humans may have occurred between 80,000 and 50,000 years ago in the Middle East. As a result, between 1-4% of the genomes of Eurasians is derived from Neanderthals.
- Neanderthals are more closely related to present-day non-Africans than to Africans. This may change the view of human origins and dispersal.
- The sequences of mtDNA fragments from 12 Neanderthal fossils indicate that three (and possibly a fourth) sub-group existed.

2. (a) Describe some problems associated with analysing DNA from fossils: _____

(b) The DNA obtained from the Denisova fossils was in excellent condition. Explain why: _____

3. (a) Explain how the Neanderthal genome project can help our understanding of the evolution of modern humans:

(b) Why might the data suggesting Neanderthals belonged to three sub-groups be regarded cautiously by some?

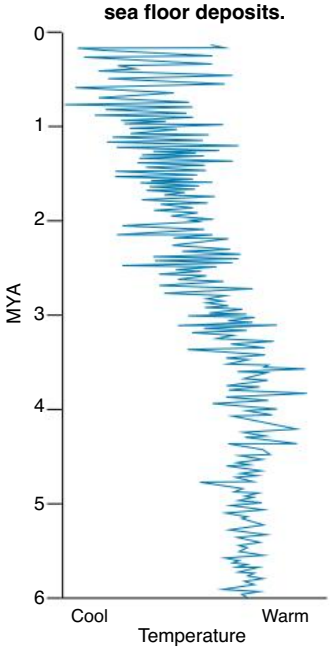
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254 The Changing Environment

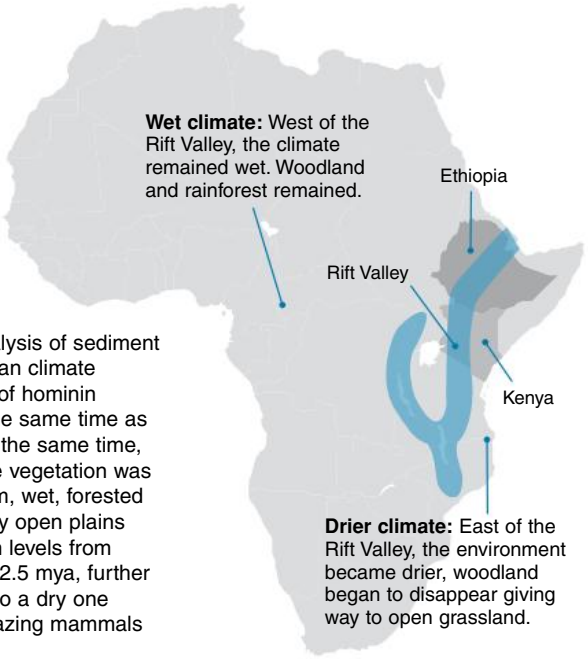
Key Idea: Understanding how both the climate and the distribution patterns of plants and animals have changed over time gives an insight into the selection pressures that may have influenced hominin evolution.

Earth's climate has fluctuated between warm and cool periods throughout its history. The fluctuations have many causes, including changes in the Earth's orbit and rotation, and changes to the Earth's surface (e.g. mountain building and continental drift). Climate change usually alters the distribution patterns of vegetation and animals.

Global temperature indicated by sea floor deposits.

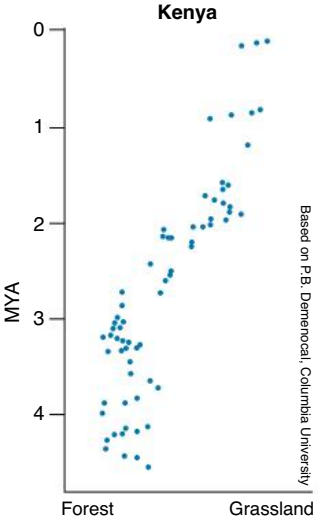


Analysis of deep sea sediment and deep ice cores provides evidence about Earth's palaeoclimate. Different depths in the core represent different periods of geological history. By analysing the isotope ratios of specific elements at different depths, it is possible to determine what the Earth's palaeoclimate was like. Evidence from these ocean cores show the Earth began cooling about 3 mya (left).

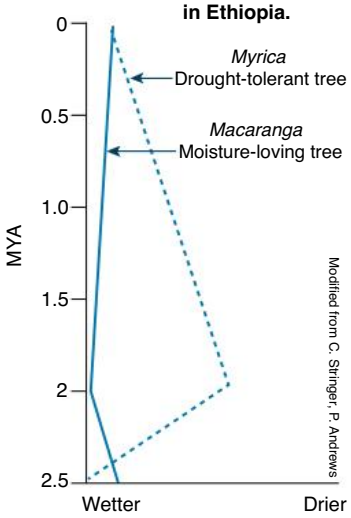


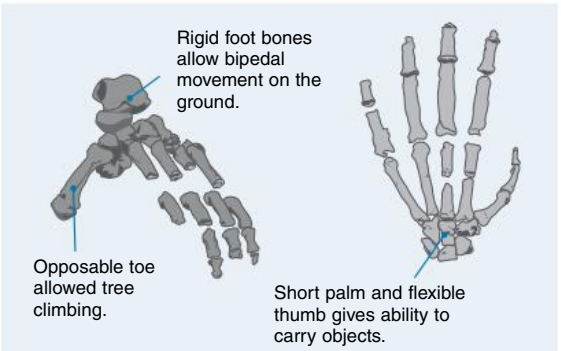
The isotope composition and pollen analysis of sediment samples from Africa show how the African climate was changing during important periods of hominin evolution. East Africa became drier at the same time as the Earth began cooling (3 mya). About the same time, carbon isotope analysis showed that the vegetation was changing from plants that favoured warm, wet, forested environments, to plants that favoured dry open plains (below, left). Pollen analysis show pollen levels from drought tolerant trees increased rapidly 2.5 mya, further indicating a change from a wet climate to a dry one (below). The fossil record shows that grazing mammals replaced browsers at about this time.

Soil carbon^{13} isotope record, Kenya



Abundance of pollen from drought/moisture tolerant trees in Ethiopia.





Fossil specimens, such as those of *Ardipithecus ramidus* (above), show that early hominins possessed features for tree climbing and for terrestrial bipedalism. The new selection pressures as the environment changed from woodland to grassland favoured terrestrial bipedalism and the evolution of the features associated with *Homo*.

1. (a) How did the African environment begin to change around 3 million years ago? _____

_____

(b) Describe the evidence that supports this change: _____

_____

_____

_____

2. Explain how the changes in the environment 3 mya may have influenced hominin evolution: _____

_____

_____

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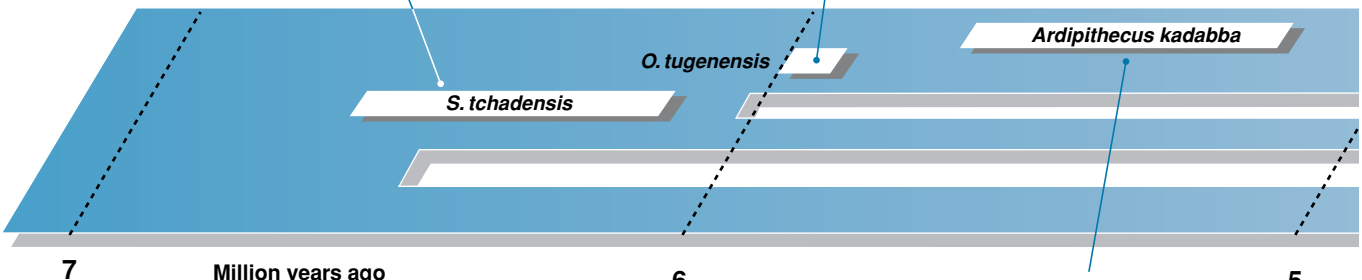
255 Hominin Evolution

Key Idea: The hominin fossil record shows clear evolutionary trends towards bipedalism, increased brain size, increased height, and reduced prognathism.

The diagram below and on the following pages shows a provisional 'consensus' view of the evolutionary timeline the hominins (modern humans and pre-humans). There is much controversy over the interpretation of fossil data from the period 4-2 million years ago (mya). Some palaeoanthropologists (scientists who study fossil hominin remains) believe that more branches existed than are shown here, with a number of adaptive radiations occurring over this period. It is almost certain that the early Australopithecines evolved into *Homo habilis*, which was ancestral to modern humans, by about 2 mya. A divergent branch, genus ***Paranthropus***, coexisted with early ***Homo***, but eventually became extinct about one million years ago. The diagram below does not attempt to show species relationships and only shows fossils that exhibit important steps in the evolution of *Homo sapiens*.

In 2001, the 6-7 my old remains of a nearly complete skull with gorilla-like features was unearthed in Chad. Nicknamed "Toumai" and assigned to a new genus, ***Sahelanthropus tchadensis***, scientists debate whether the skull's features place it in the human family tree, or whether it represents the remains of a proto-gorilla.

The 6 my old remains of five chimpanzee-sized ***Orrorin tugenensis*** specimens were unearthed at Baringa in Kenya in 2000. The teeth are very humanlike and a perfectly preserved thigh bone clearly shows features associated with walking upright (bipedalism).



New DNA and biochemical evidence suggests that the **last common ancestor** of hominins and apes occurred between 5 and 10 million years ago. The last common ancestor should have a combination of features reminiscent of both humans and apes.

Ardipithecus kadabba is dated at 5.8 my old and is known only from a handful of bones consisting of teeth and parts of the skeleton.

	S. tchadensis	*O. tugenensis*
Skull	 Prognathic. Heavy brow ridges. Small teeth with thick enamel	 Relatively small teeth, with thick enamel. Canines ape-like.
Brain	Brain volume: 360-370 cc (cubic centimetres)	Brain volume: Unknown
Body	Height around 1 m. Weight unknown. Mostly tree living, but could probably walk upright for short distances.	Part of femur indicates it may have been bipedal.

1. Distinguish between **hominins**, **hominids** and **hominoids**: _____

2. (a) Describe the key identifying features of the (gracile) **australopithecines**: _____

   (b) Which species are normally assigned to this group? _____

3. (a) Which species is considered to be the common ancestor to later australopithecines and also to genus *Homo*?

   (b) State the date range for the hominin you named in 3(a): _____

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What's the Difference?

Some of the terms used to describe humans and their ancestors and relatives sound very similar and are often confused.

The **hominoids** constitutes the superfamily to which the hominins and all the apes belong.

A **hominid** includes the hominins and the great apes (e.g. chimpanzees).

A **hominin** is a primate that is habitually bipedal e.g. humans and their immediate ancestors.



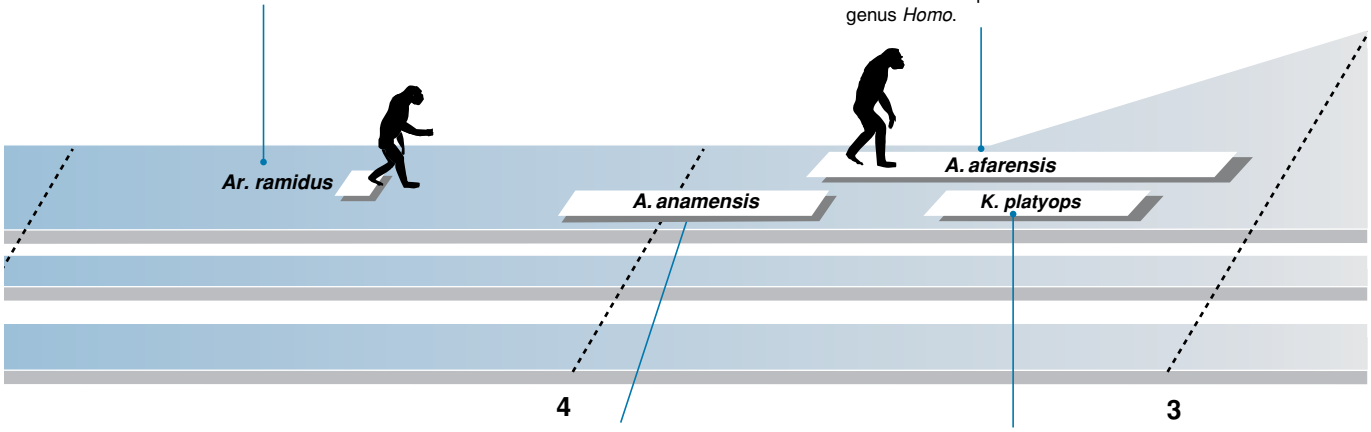
Sahelanthropus tchadensis

Australopithecines

The earliest of these hominins were among the first apes to achieve bipedalism. They possessed a **gracile** body form that was lightly built with fine bone structure and were probably opportunistic omnivores, scavenging meat from carcasses and feeding off a wide range of resources.

Ardipithecus ramidus was an early hominin. Fossils suggest that it was at least partially bipedal with teeth that were also more humanlike. *Ar. ramidus* is dated at 4.4 my old).

Australopithecus afarensis is thought to be the common ancestor to both Australopithecines and the genus *Homo*.



Australopithecus anamensis is dated at about 4 million years old. Over one hundred fossil specimens representing 20 indivuals have been found in Kenya and Ethiopia.

Kenyanthropus platyops, a 3.5 my old hominin, coexisted with *A. afarensis*.

Ar. ramidus ramidus



Prognathic. Large brow ridges. Relatively advanced hominid teeth.

Brain volume: 300-350 cc

Height 1.2 m. Weight 50 kg. Adapted for tree living but had many features which clearly point to an ability to walk upright.

K. platyops



Large cheek bones, producing a flat face.

Brain volume: 450 cc

Height unknown. Weight unknown.

A. afarensis



Prognathic. Large brow ridges. Relatively large incisors

Brain volume: 375-550 cc

Height 1.5 m (male), 1 m (female). Weight 50 kg (males heavier than females) Analysis of leg and hip bones as well as fossil footprints show *A. afarensis* was bipedal.

4. State the date range palaeoanthropologists believe the hominini and chimpanzees last shared a common ancestor:

_____

5. (a) Which human evolutionary trend appeared to begin developing first - an increase in brain size or bipedalism?

_____

(b) When did evidence of this trend first appear? _____

(c) Which species above can we be almost certain was bipedal? _____

(d) What is the evidence for this? _____

_____

_____

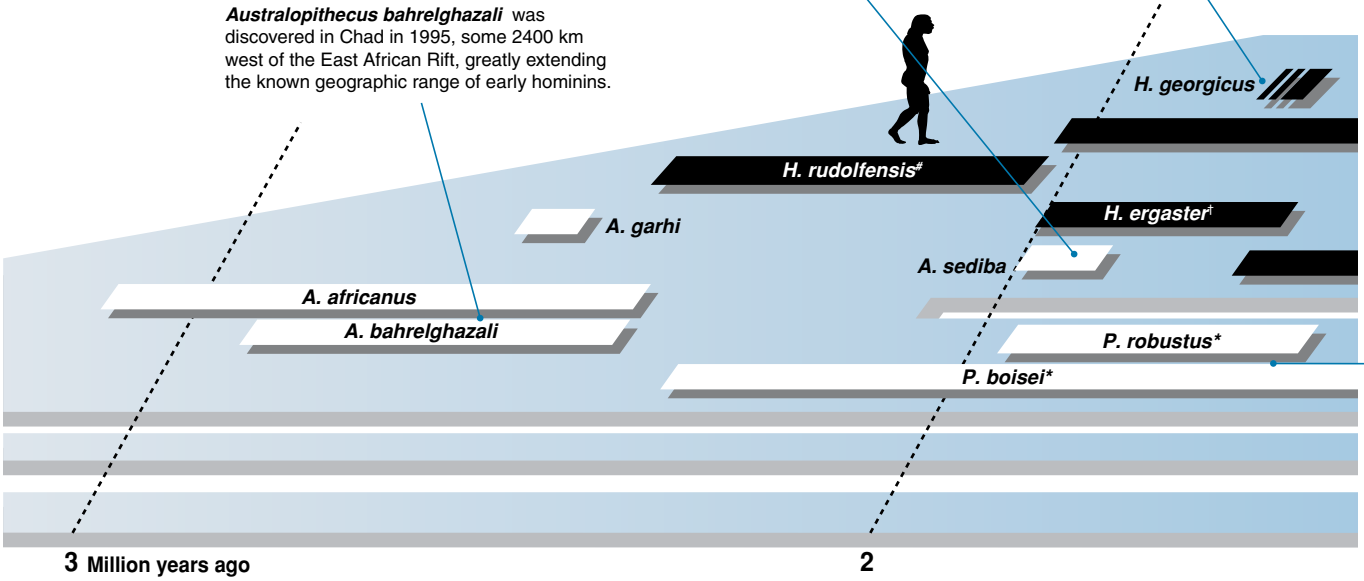
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Small-brained fossils, found at **Dmanisi** and dated at 1.8 million years old, have been assigned as *Homo georgicus*. Their location in the former Soviet republic of Georgia, suggests an unexpectedly early hominin exodus from Africa.

The discovery of ***Australopithecus sediba*** has raised questions over the later evolution of the australopithecines.

Australopithecus bahrelghazali was discovered in Chad in 1995, some 2400 km west of the East African Rift, greatly extending the known geographic range of early hominins.



Paranthropines (*)

These early hominins represent a group specialised for eating a bulky, low-grade vegetarian diet. They developed large cheek teeth, powerful chewing muscles and a generally **robust** skull (large crests for muscle attachment, heavily buttressed face).

Habilines (#)

This group shows the first signs of brain enlargement, more meat in the diet as well as the first recognisable stone tool culture. The post-cranial (below the head) skeleton remains small and slight much like that of the australopithecines.

	A. africanus	*P. robustus*
Skull	 Reduced brow ridges. Higher forehead. Small canines and large molars	 Pronounced sagittal crest. Large flat face. Brow ridges. Massive jaw and molars.
Brain	Brain volume: 450 cc	Brain volume: 530 cc
Body	Height: 1.3 m (male larger than female). Weight: 40 kg. Probably capable of upright walking, but spent time climbing and on all fours.	Height: 1.3 m. Weight: 40 kg. Males larger than females. Possibly bipedal, but mainly quadrupedal.

2 Million years ago

6. (a) What is a **megadont**? _____

   (b) Identify two species belonging to this group: _____

   (c) Suggest why they may have become extinct: _____

   _____

7. Using the diagram above determine:

   (a) The total number of hominin species (i.e. from 6 mya to the present day): _____

   (b) The number of hominin species that existed between 3 and 2 million years ago: _____

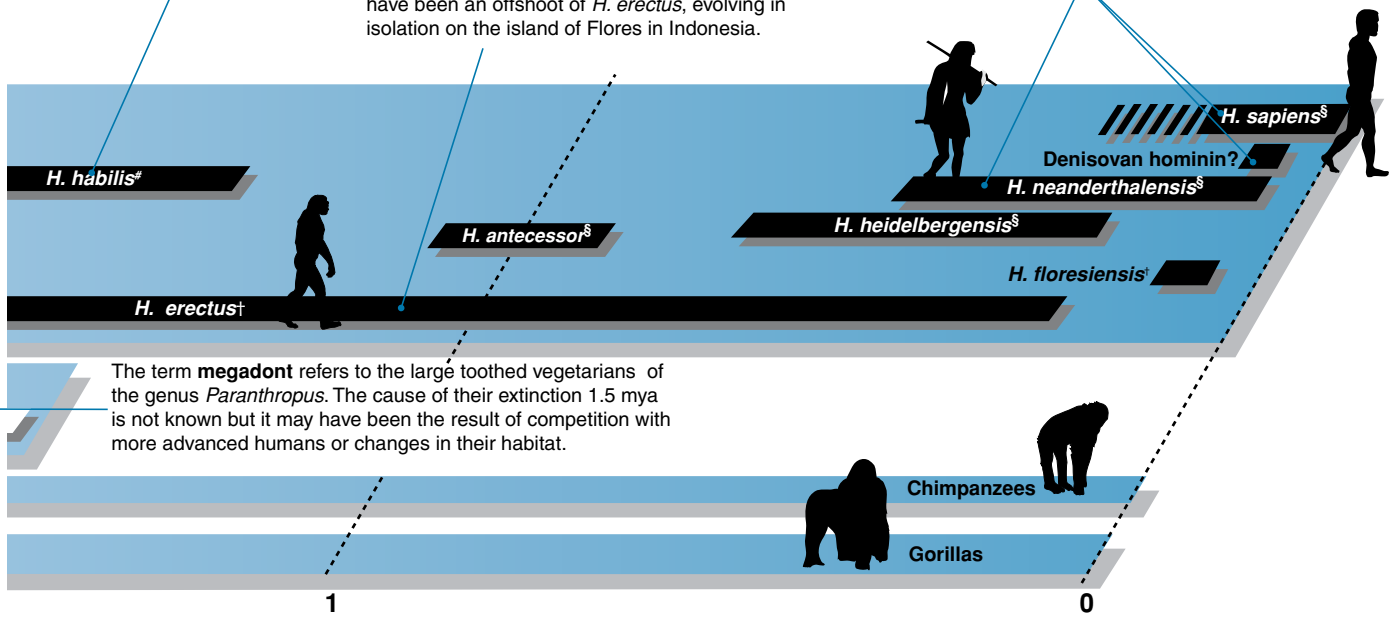
   (c) At what date did the *Homo* genus first appear? _____

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Homo habilis species may be too variable to be considered a single species and should be split into two. The additional and older species is ***Homo rudolfensis***. Only one of these hominins may have been our direct ancestor.

Homo erectus was a long lived species thought to be the first to venture out of Africa. The small hominin *H. floresiensis* appears to have been an offshoot of *H. erectus*, evolving in isolation on the island of Flores in Indonesia.

Modern humans may have appeared more than 160,000 years ago. **Neanderthals** were not the direct ancestors of modern humans. Recent comparison of DNA suggest that Neanderthals split from the human lineage as far back as 600,000 years ago. Recent finds in Russia of a tooth and a finger bone have been assigned to a **Denisovan hominin**. The finds are dated at older than 30,000 years and are genetically distinct from both Neanderthals and modern humans.



The term **megadont** refers to the large toothed vegetarians of the genus *Paranthropus*. The cause of their extinction 1.5 mya is not known but it may have been the result of competition with more advanced humans or changes in their habitat.

Erectines (†)

Big changes in the post-cranial skeleton with body height achieving modern proportions. Marked behavioural changes are matched by increasing brain volume, with sophisticated tool manufacture and use employed to kill and process small sized game. It is still unclear whether *H. floresiensis* belonged to this group.

Archaic and Modern Humans(§)

Rapid advances in brain size are associated with a suite of new behaviours during this period. Anatomically modern humans emerge from one of the many regional variants and rapidly spread through much of the Old World.

H. habilis	***H. erectus***	***H. neanderthalensis***	***H. sapiens***
 Little prognathism. Parabolic dental arcade. Reduced canines and molars.	 Little prognathism. Prominent brow ridge. Parabolic dental arcade. Reduced teeth.	 Prominent brow ridge, sloping forehead, and occipital bun. Flat nose.	 High forehead and domed skull. Prominent chin. Highly reduced teeth and jaw.
Brain volume: 500-800 cc	Brain volume: 1000-1250 cc	Brain volume: 1200-1700 cc	Brain volume: 1350 cc
Height: 1.3 m (male larger than female). Weight: 40 kg. Bipedal. Associated with Oldowan tools	Height: 1.7 m. Weight 60+ kg Fully bipedal. Associated with Acheulian tools.	Height: 1.7m. Weight 65+ kg Heavy set skeleton and body adapted to a physically demanding environment. Associated with Mousterian tools.	Height: 1.75m. Weight: 65+ kg. Legs longer than arms. Gracile skeleton. Associated with highly sophisticated tool cultures starting with upper Paleolithic tools.

1 0

(d) Identify the extant (living) *Homo* species: _____

8. Explain why it is most unlikely that the Neanderthals were ancestors to modern humans: _____

_____

_____

_____

9. (a) What is the evolutionary trend in hominin height? _____

_____

(b) Identify the tallest hominin: _____

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256 The Origin of Modern Humans

Key Idea: The two main contesting theories on the origin of modern humans are the multiregional hypothesis and the replacement (out-of-Africa) hypothesis.

Two primary hypotheses have been put forward to explain where modern humans evolved and what happened to the *Homo* species that preceded them. The multiregional hypothesis proposes that modern humans evolved from *Homo erectus* everywhere that *Homo erectus* preceded them. The replacement hypothesis proposes that modern humans evolved from an African *Homo erectus* (via an intermediate species called *Homo heidelbergensis*) and replaced all other forms of *Homo* throughout the Old World.

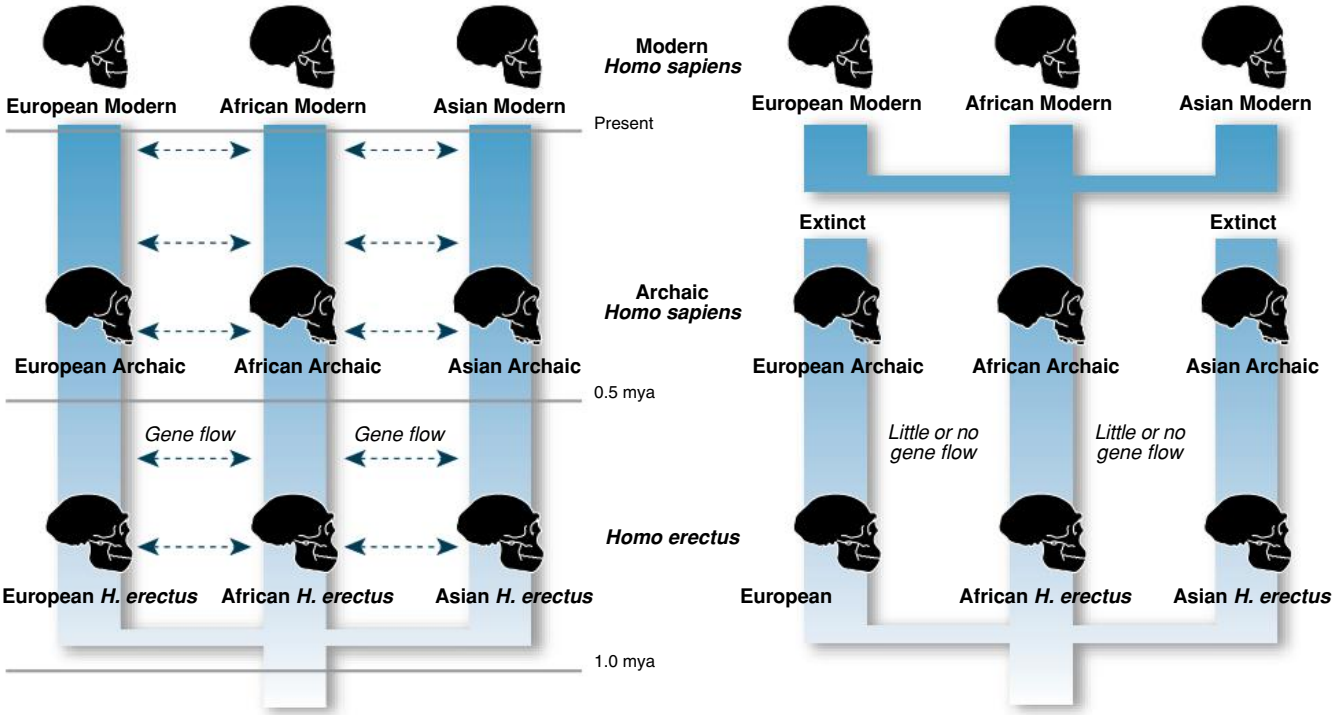
Multiregional Model

In this model, modern Homo sapiens emerged gradually throughout the world and, as the populations dispersed, they remained in 'genetic contact'. This gene flow between neighbouring populations ensured that the general 'modern human blueprint' was adopted by all. This limited gene flow still allowed for slight anatomical differences to be retained or develop in the regional populations. This model is based largely on fossil evidence and the anatomical characteristics of modern populations, but advocates of the model maintain that the mitochondrial DNA data can be interpreted in a way that supports it.

See ***Scientific American*** **SPECIAL ISSUE: New Look at Human Evolution,** Vol. 13(2), 2003, pp. 46-61, for two excellent articles on each of these models.

Replacement Model

Also known as the "Out of Africa Hypothesis" and "Eve Hypothesis". This model states that modern humans evolved from archaics in one location, Africa, and then spread, replacing the archaic populations, without interbreeding. Modern human variation is thus a relatively recent phenomenon. Mitochondrial DNA (mtDNA) analysis of modern endemic human populations showed that the highest level of genetic variation in mtDNA occurs in African populations, implying that *H. sapiens* arose first in Africa. Using the genetic distance between African populations and others as a measure of time, this model places the origin of modern humans at back to a single female (Eve) who lived there some 200,000 years ago.



Predictions made by this model

1. Fossils that show the change from one stage to the next in all geographic regions (transitional forms).
2. Modern traits should appear in the fossil record somewhat simultaneously all over the Old World range of Archaic *Homo sapiens*.
3. Today's modern "racial" traits characteristic of a particular region can be traced back to ancient forms in that region.
4. The human species today should have a high degree of genetic diversity since it is an old species with distinct populations that have had a lot of time to accumulate genetic differences.
5. The amount of genetic variation within each modern human group is about the same since they have all been evolving together.

Predictions made by this model

1. Transitional forms would be found in only one place (in this case Africa) which is the area of origin for modern humans.
2. Modern traits should appear first in one location (Africa) and then later elsewhere as the modern population spread to other parts of the Old World.
3. Modern and archaic populations should overlap in time outside the area that moderns originated (the process of replacement would not be instantaneous).
4. Humans today should have relatively little genetic diversity since the species is young.
5. Today's modern populations should differ in the amount of genetic variation, the most diversity being found in the region where moderns first evolved (this would have been the oldest group and therefore the one with the most time for mutations to accumulate).

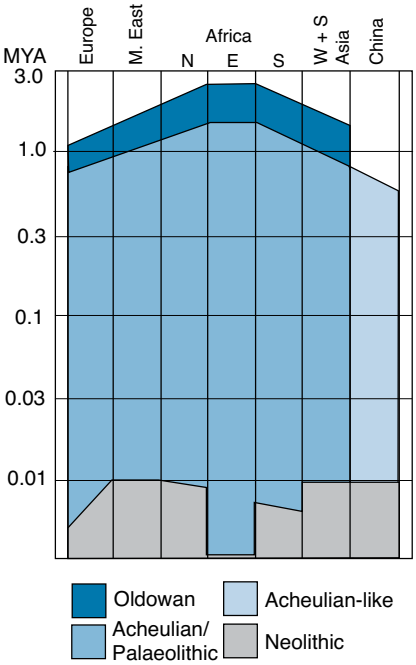
Source: Michael A. Park, Biological Anthropology, Mayfield Publishing, 1996

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Evidence for Dispersal

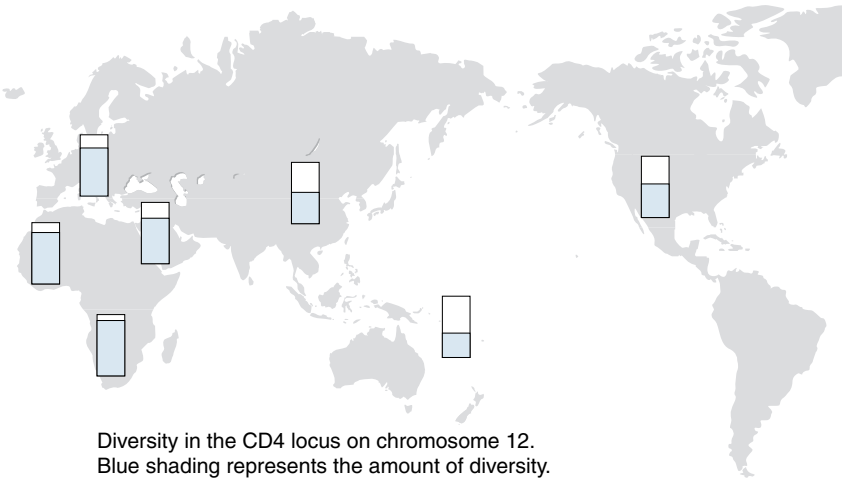
There is little doubt that human ancestors evolved in Africa. Fossils of ancient hominins such as *Australopithecus afarensis*, *A. africanus* and *Homo habilis* are all found in Africa. Analysis of fossils and genetic evidence supports theories that *Homo erectus* migrated out of Africa about 1.8 mya. Populations of *H. erectus* remaining in Africa gave rise to *H. heidelbergensis*, which also migrated out of Africa about 650,000 years ago but remained in Europe and Western Asia. Those populations gave rise to *H. neanderthalensis*. *Homo heidelbergensis* in Africa eventually gave rise to *H. sapiens*. *H sapiens* began to migrate out of Africa between 120,000 and 80,000 years ago but the movement was halted by a period of cold climatic conditions. Around 60,000 years the final migration of modern humans out of Africa and across Europe and Asia began.

Acheulian stone tools are found throughout Africa, Europe, and South and Western Asia, but appear to be largely absent from Eastern Asia. Tool cultures develop first in Africa then move to other parts of the world.



Analysis of various loci in the human genome show humans have very little genetic diversity. The analysis shows that outside of Africa, human genetic diversity is a sub-set of African genetic diversity.

	Diversity (0 = low)		
Locus (measurement)	**Africa**	**Asia**	**Europe**
30 microsatellites	0.807	0.685	0.730
Xq 13.3 (short arm X chromosome)	0.035	0.025	0.034
50 autosomal sequences	0.115	0.061	0.064
mt DNA control region	2.08	1.75	1.08



Diversity in the CD4 locus on chromosome 12.
Blue shading represents the amount of diversity.

1. State at what date the two models suggest that anatomically modern humans shared a common ancestry:

   (a) Replacement model: _____ years ago (b) Multiregional model: _____ years ago

2. (a) Which hominin was the first to migrate out of Africa? _____

   (b) At about what date did this migration occur? _____

3. How many successive waves of *Homo* migrated out of Africa? _____

4. Explain why it is unlikely that Neanderthals ever interbred with modern humans to any great degree:

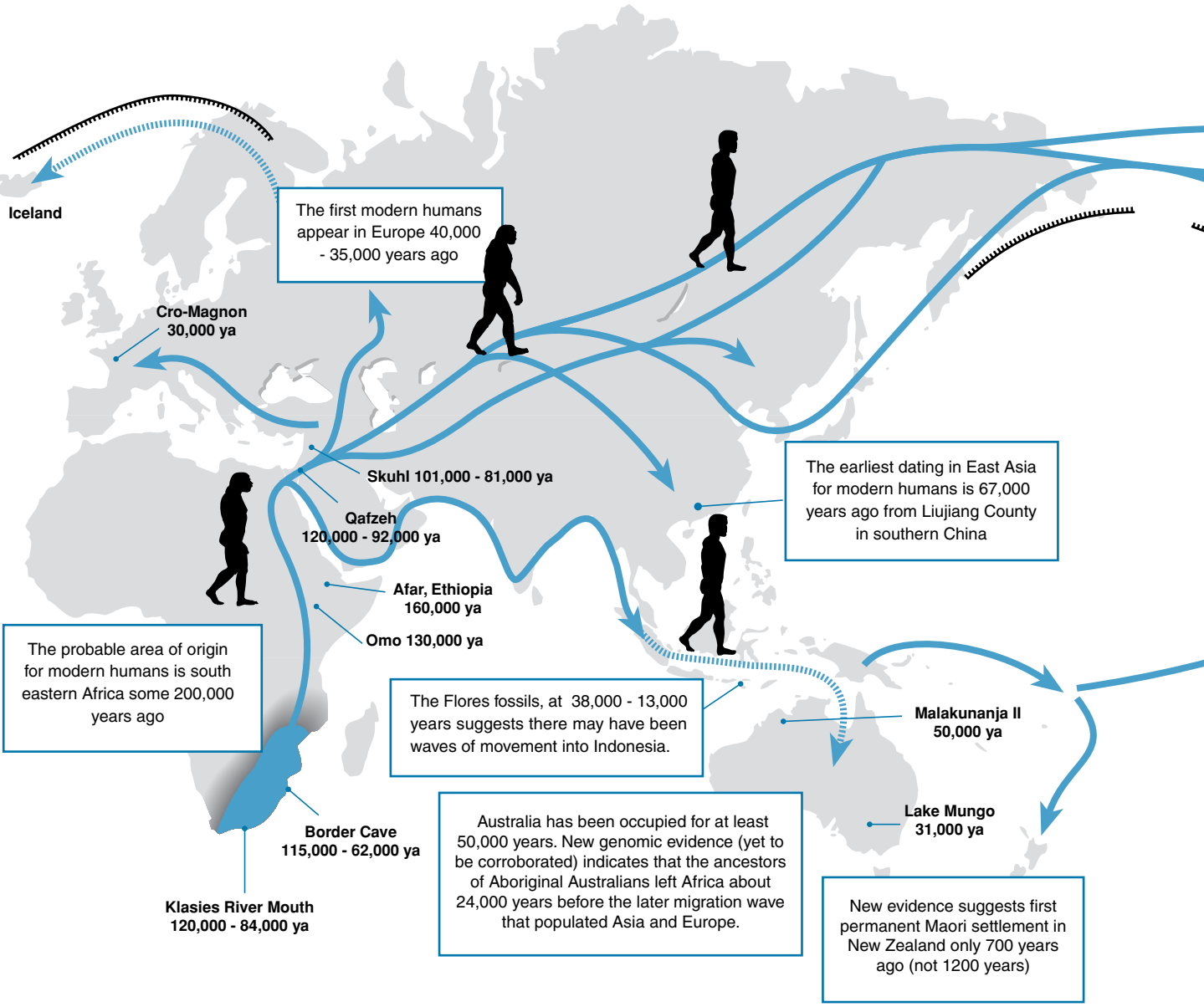
5. Summarise the evidence above and decide which hypothesis it supports: multiregional or out-of-Africa:

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257 The Dispersal of Modern Humans

Key Idea: Hominins evolved in Africa. Around 60,000 years ago *Homo sapiens* began the final hominin migration out of Africa, eventually reaching all parts of the world.

The map below shows a suggested probable origin and dispersal of modern humans throughout the world. An African origin is almost certain, with south eastern Africa being the most likely region. The dispersal was affected at crucial stages by the last glacial, when Ice sheets covered much of Europe. Dispersal was also affected by the presence or absence of 'land bridges' formed during the drop in sea level that occurs with the onset of glacials. Recent evidence suggests that island-hopping and coastal migration may also have been important, e.g. for the movement of people into Indonesia. The late development of boating and rafting technology slowed dispersal into Australia and the Pacific. New Zealand was one of the last places on Earth to be populated. (On the map, **ya** = years ago.)



1. (a) Study the map above and suggest why humans reached Australia before they reached Western Europe, even though Australia is further away from their point of origin:

_____

_____

_____

(b) Suggest why New Zealand was one of the last land masses populated by humans: _____

_____

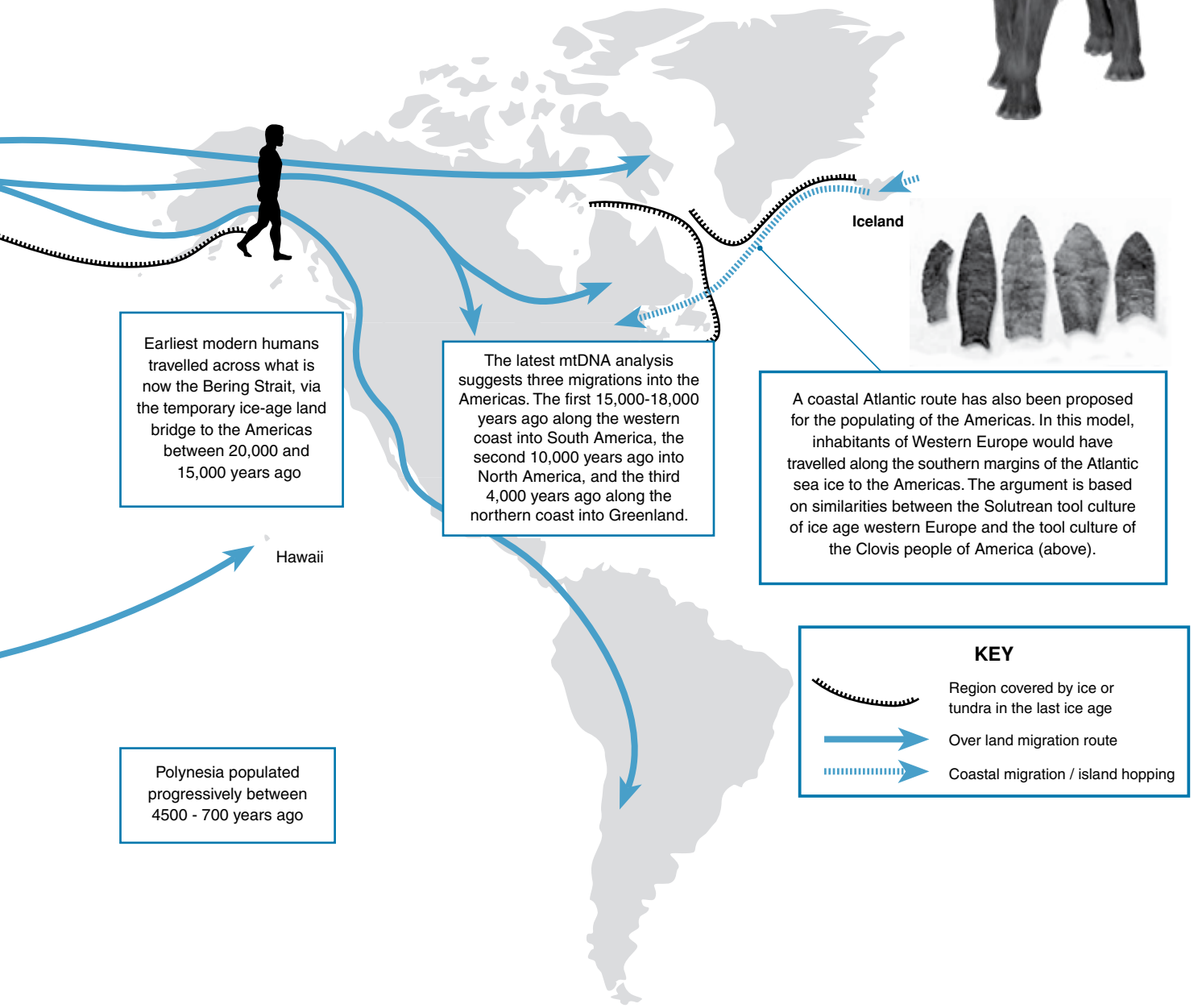
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The movement of humans into new parts of the world has often been cited as the major reason for the extinction of the many large animals that lived around 40,000 to 15,000 years ago, such as the woolly mammoth. However, new evidence suggests climate change near the end of the last glaciation was the major factor in the extinction of these animals. Also, good evidence of mammoth hunting is relatively difficult to find, suggesting humans did not play a big part in their extinction.





2. Using examples, discuss the importance of land bridges and glaciations in the global dispersal of modern humans:

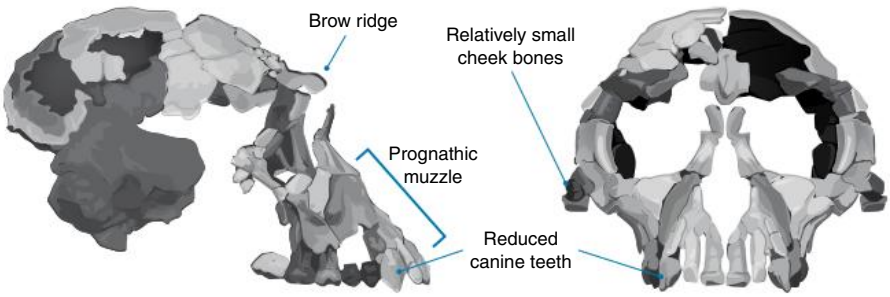
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258 The Importance of *Ardi*

Key Idea: Analysis of the skeleton of *Ardipithecus ramidus* has produced findings that suggest bipedalism and a manipulative hand are very ancient features.

The first fossils of *Ardipithecus ramidus* were discovered in the Middle Awash region of northeastern Ethiopia in 1994. After many years of excavation, a partial skeleton was unearthed. Studying the skeleton of *Ar. ramidus* is beginning to change our understanding of hominin evolution. Until recently, it had been theorised that our earliest ancestors moved about very much like the chimpanzees of today. However the evidence from the *Ar. ramidus* skeleton shows that this is not the case and that bipedalism developed in quite a different way to what was once thought. Moreover, a dextrous hand developed early and is also an ancient trait.

Skull: The skull of *Ar. ramidus* shares certain features with *Australopithecus*, including a reduction in the size of the canine teeth in both male and females. This implies a reduction in aggression between males. The orientation of the base of the skull on which the brain stem rests suggests that the parts of the brain involved in visual and spatial perception were already beginning to develop.

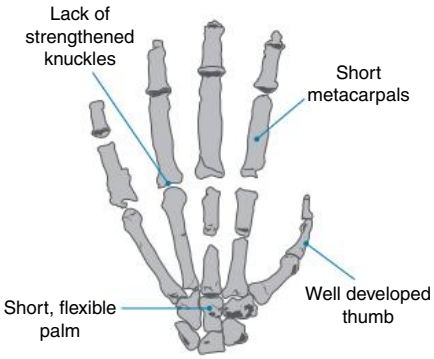


***Ardipithecus ramidus* skull features**

Hand: Analysis of the hand of *Ar. ramidus* shows that it is similar to our own, and that human hands are therefore close to the primitive form and not as greatly modified for tool use as was previously thought. *Ar. ramidus* had a flexible wrist and the opposable thumb was well developed. By contrast, chimpanzees move on the ground by knuckle walking, a motion that requires strengthening of the wrist and knuckle bones and lengthening of the palm, making the hand less flexible and not as dextrous.



Human hand



The primitive features of the ***Ardipithecus* hand** that are shared with *Homo*.

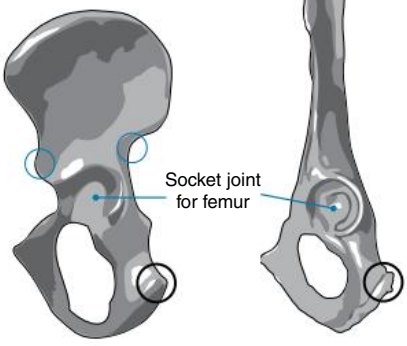
Pelvis: The pelvis of *Ar. ramidus* indicates that the modern pelvis, evolved for bipedal locomotion, began its evolution in the trees. Although several features of the upper pelvis strongly indicate bipedalism, features of the lower pelvis show that muscles associated with tree climbing were still well developed.

Features associated with bipedalism that are shared by *Ardipithecus* and *Homo*

Feature associated with tree climbing that is shared by *Ardipithecus* and *Pan*



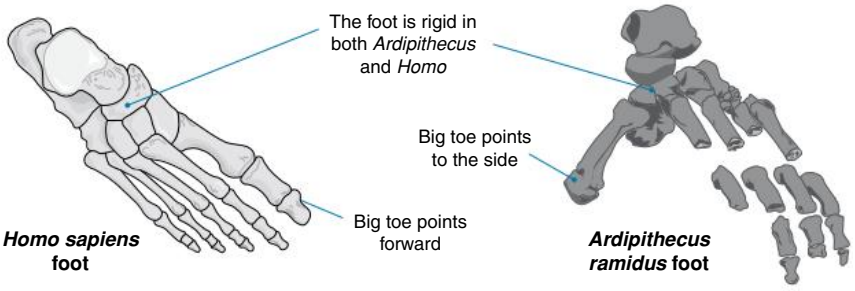
Human pelvis



***Ardipithecus ramidus* pelvis**

Chimpanzee pelvis

Foot: The foot of *Ar. ramidus* is a generalised one, with some human-like features, such as a rigid foot, as well as some modern ape-like features, such as an opposable big toe. These features indicate that *Ar. ramidus* spent considerable time climbing in trees.



***Homo sapiens* foot**

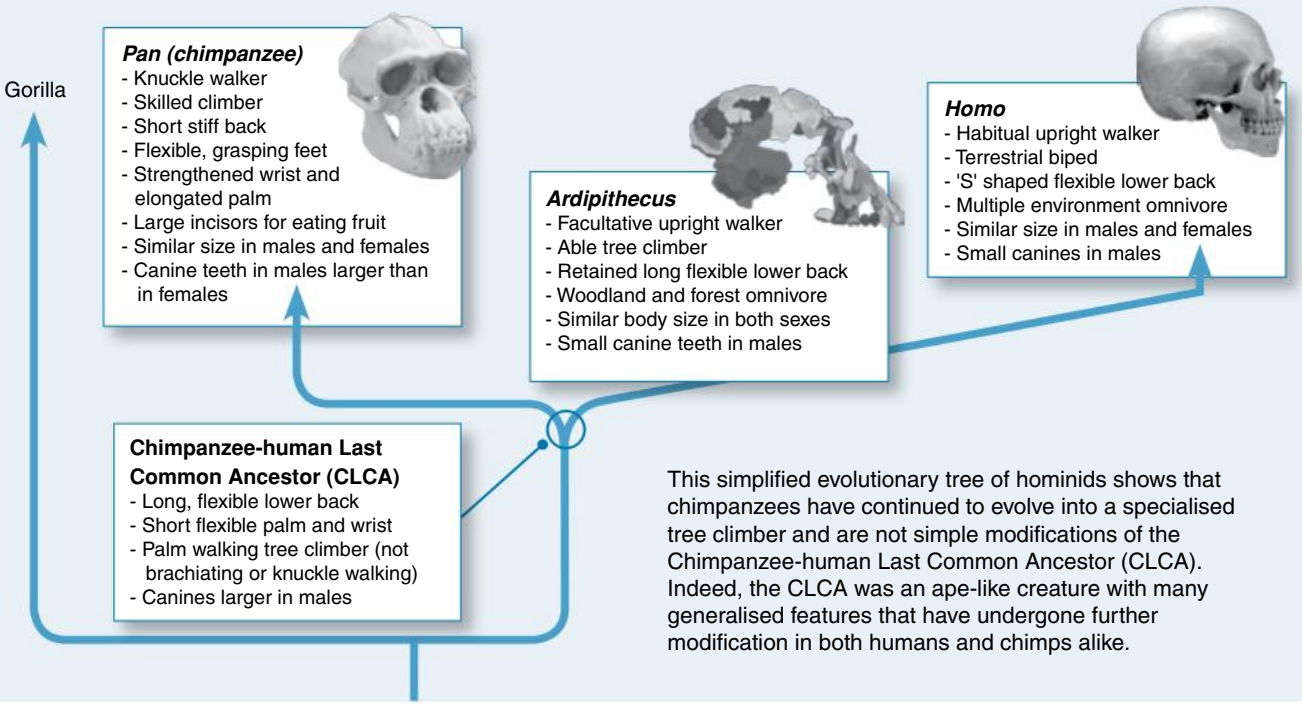
***Ardipithecus ramidus* foot**

Images redrawn from C. Owen Lovejoy, et al Science, vol 326, 2009

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Proposed evolutionary tree with *Ardipithecus*



This simplified evolutionary tree of hominids shows that chimpanzees have continued to evolve into a specialised tree climber and are not simple modifications of the Chimpanzee-human Last Common Ancestor (CLCA). Indeed, the CLCA was an ape-like creature with many generalised features that have undergone further modification in both humans and chimps alike.

Modified from C Owen Lovejoy, et al Science, vol 326, 2009

1. Describe the evidence for reduced aggression between *Ar. ramidus* males: _____

2. Explain why the human hand might now be viewed as the primitive type: _____

3. Describe the evidence for bipedalism developing in a primarily arboreal (tree-dwelling) ancestor rather than in a knuckle walking, terrestrial ancestor:

4. How does having a generalised body plan increase possible evolutionary pathways? _____

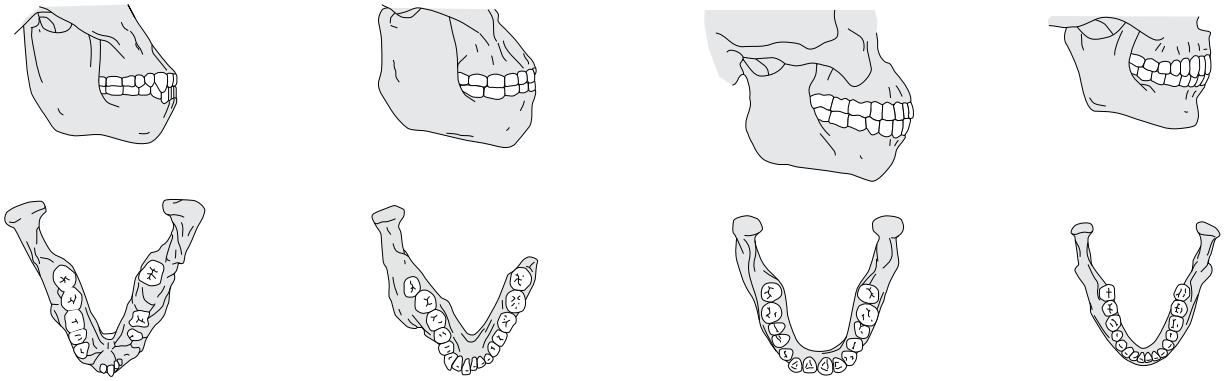
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259 Trends in Dentition

Key Idea: Changes in dentition (the type, number, and arrangement of teeth) and jaw structure of our hominin ancestors can reveal information about our evolution.

During early hominin evolution teeth (especially the molars) and jaws tended to be large. The paranthropines are the extreme example of this trend. Their diet of coarse vegetation required very large and powerful jaws and molars. During the course of hominin evolution, there was a general trend for a reduction in the size of the teeth tooth and jaw. This was a likely consequence of including a greater proportion of cooked foods, which required less chewing, in the diet. The teeth of modern humans are relatively small and generalised, reflecting an omnivorous diet of mainly processed (e.g. cooked) foods.

Early Hominins → **Late Hominins**



Australopithecus afarensis

- Relatively large canine teeth
- Relatively large jaw
- V-shaped dental arcade
- Thin tooth enamel
- Diet probably consisted of fruits with some tougher material

Australopithecus africanus

- Reduced canine teeth
- Large molars
- Dental arcade intermediate between *A. afarensis* and *H. sapiens*
- Thick tooth enamel
- Diet probably included vegetable matter, nuts, seeds, insects, and eggs

Homo erectus

- Thick jaw bones
- No chin
- Relatively large molars
- Parabolic dental arcade
- Thick tooth enamel
- Diet probably included vegetable material and a large proportion of meat

Homo sapiens

- Shortened jaw, allows large bite force to be generated with little effort
- Chin reinforces jaw, but leaves room for tongue muscles
- Thick tooth enamel
- Small molars adapted to chewing cooked and soft food
- Parabolic dental arcade

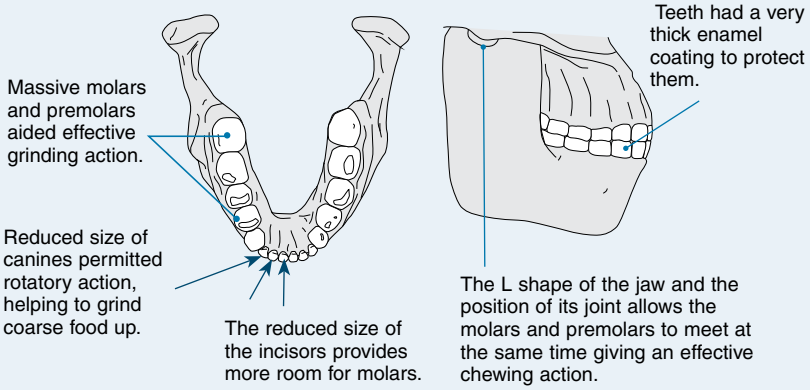
Dental formulae all follow: I-2, C-1, P-2, M-3



In many primates, the canine teeth are used in behavioural and social interactions, especially in species which show marked sexual dimorphism. Threat gestures, such as yawning (above), help maintain social order.

Adaptations to a Coarse Diet

Paranthropus boisei had jaws and teeth adapted to a diet of coarse vegetation and hard seeds. Their jaws produced a massive bite force of 2161 Newtons, which helped to break food up.



1. Describe the general trend in the evolution of hominin teeth: _____

_____

_____

_____

2. What is one possible purpose of the chin in modern humans? _____

_____

_____

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260 Bipedalism and Nakedness

Key Idea: Bipedalism provided advantages such as better provisioning, more efficient thermoregulation, and an improved view of the environment.

The first major step in the development of humans as a distinct group from apes was their ability to adopt the habitually upright stance we call **bipedalism**. Closely linked to this shift was the reduction in body hair. A number of selection pressures for hair reduction are described in the left column below. Early studies suggested that bipedalism and hair reduction were both evolutionary responses to the changing climate of East Africa about 7-3 million years ago. However, recent (2009) analysis of the 4.4 million year old *Ardipithecus* fossils indicates that these very early hominins were still primarily forest dwellers, so any current hypotheses must account for the emergence of bipedalism in a forested environment. The *Ardipithecus* finds indicate that bipedalism was strongly associated with provisioning, and was later reinforced by a move into less forested habitats as savannah became established throughout Africa during the later Miocene.

Hair Reduction

Retention of head hair

Hair on the head (and to a lesser extent the shoulders) has been retained to reflect and radiate heat before it reaches the skin of the exposed part of the body.

Parasite control

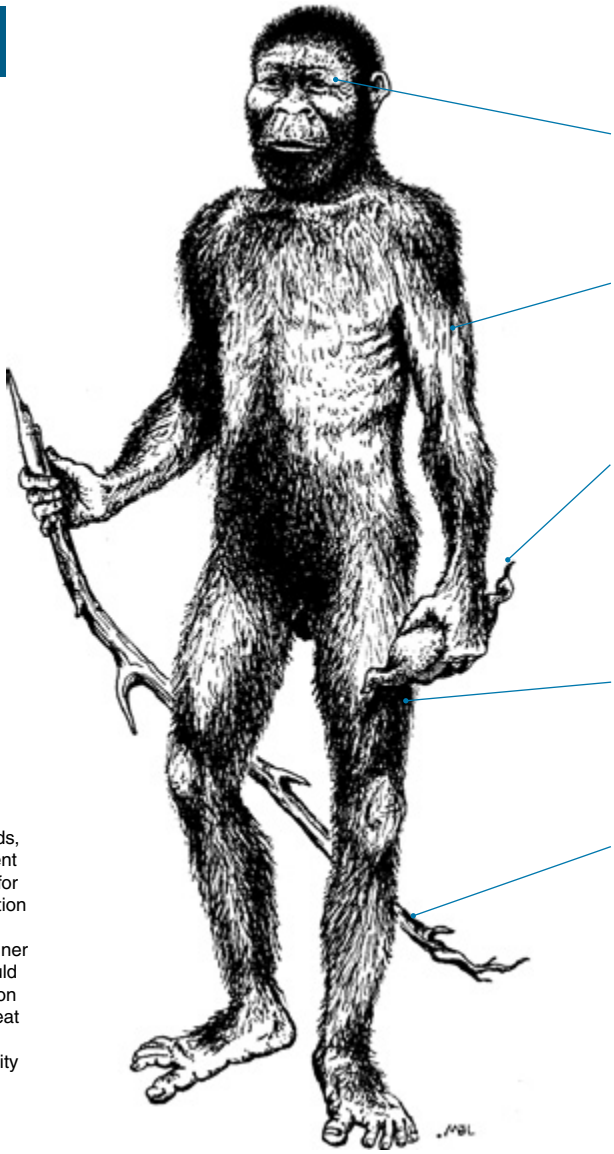
A reduction in body hair would have made the control of parasites such as fleas, ticks and lice much easier. This became particularly important when early hominins began to use a 'home base' rather than continually wandering. Parasites, such as fleas, need to complete their life cycle at a single location so that hatching eggs can reinfect their host.



Lice Ticks Fleas

Thermoregulation

About 3 mya, the vegetation patterns in East Africa began to favor open grasslands, with fewer forested areas. This environment would have provided fewer opportunities for shelter from the hot sun, creating a selection pressure for the refinement of several thermoregulatory mechanisms. Shorter, finer hairs (not hair loss) in early hominins would have allowed greater heat loss via radiation from the skin surface. Well developed sweat glands in humans enable heat loss at an astounding 700 watts m^2 of skin (a capacity not approached by any other mammal).



Bipedalism

Seeing over the grass

An upright posture may have helped early hominins to see predators or locate carcasses at a distance.

Carrying offspring

Walking upright enabled early hominins to carry their offspring, facilitating movement as a family group.

Provisioning

The ability to carry food while walking appears to have been a major selective pressure in the initial development of bipedlism. Potential mates would have favoured males with the ability to provide energy-rich foods. The capacity to carry food away from a kill site or growing site to a position of safety would have had great survival advantage.

Efficient locomotion

Once bipedalism was established, changing habitats would have reinforced its further development. Bipedal motion provided an efficient means of moving across the growing expanses of savannah.

Holding tools and weapons

Tool use was probably a consequence of bipedalism, rather than a cause. Upright walking appears to have been established well before the development of hunting in early hominids.

Thermoregulation

Upright walking exposes 60% less surface area to the sun at midday and there is greater air flow across the body when it is lifted higher off the ground.

1. Discuss the selection pressures that are likely to have been important in the initial development of bipedalism, given that the current evidence indicates it arose in a forested environment:

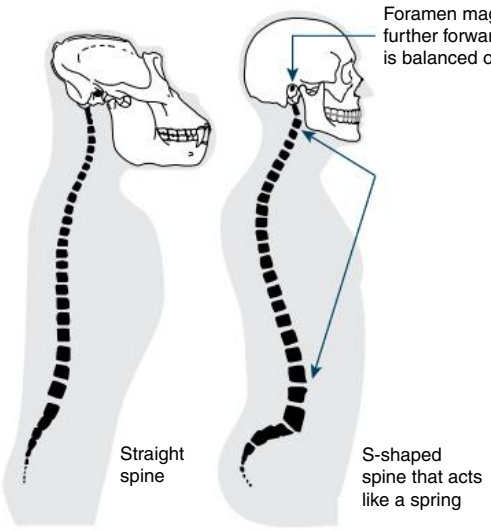
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261 Adaptations for Bipedalism

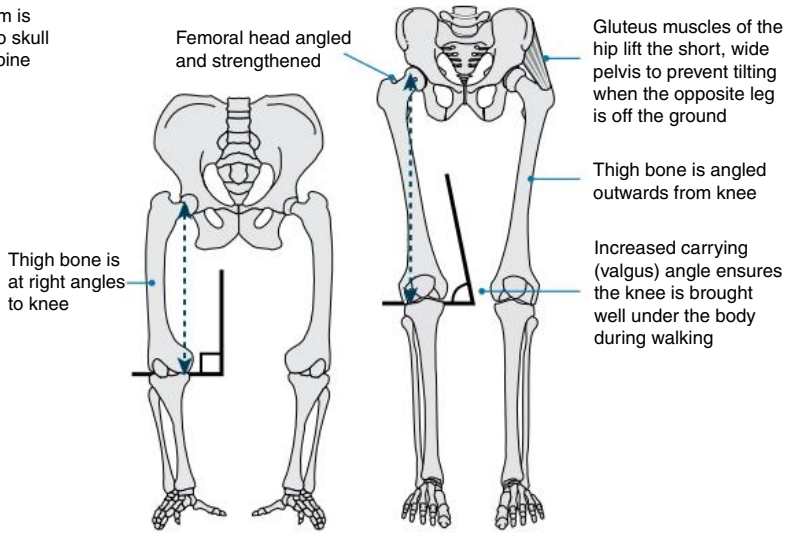
Key Idea: Important changes in the skeleton are associated with the move to bipedal locomotion in early hominins.

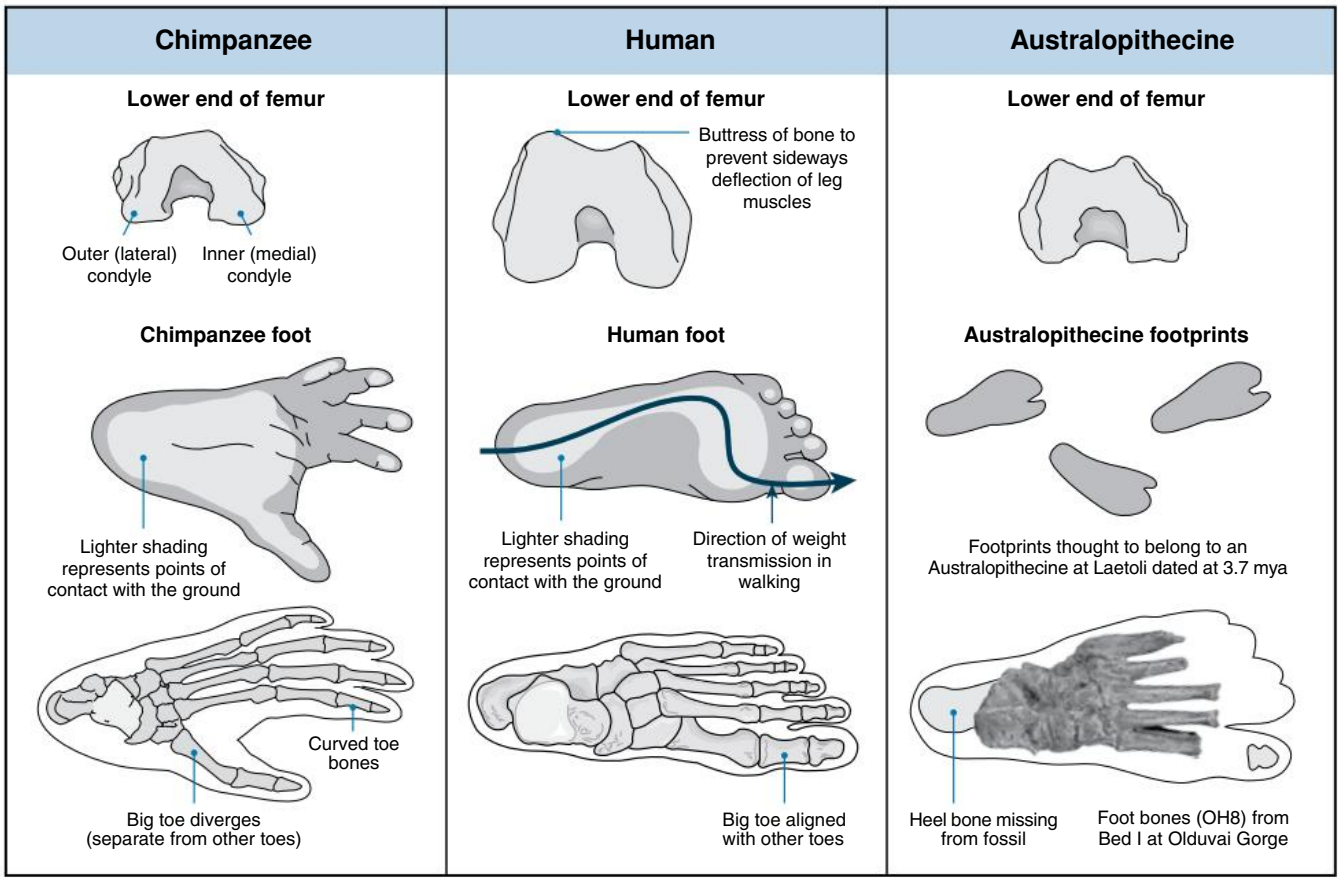
The reconstruction of '**Lucy**' (*Australopithecus afarensis*) pictured on the next page illustrates skeletal features in an early bipedal hominin. While Lucy still possessed some ape-like characteristics, she was a fully-bipedal hominin with all the adaptations associated with bipedal locomotion. Lucy was small at only 1.1 m tall. Although there is no doubt that she was habitually bipedal, a number of skeletal features suggest that tree climbing was still an important part of this hominin's niche. Such activities may have been associated with escape, security while sleeping, and foraging. *A. afarensis* forms an important link between the quadrupedal locomotion of apes and bipedalism in hominins.

Gorilla spine **Human spine**



Gorilla legs **Human legs**





1. Referring to the diagram above, describe whether each of the **australopithecine fossils** compare more closely to the *chimpanzee* or *human* examples (i.e. to which do they bear the closest resemblance):

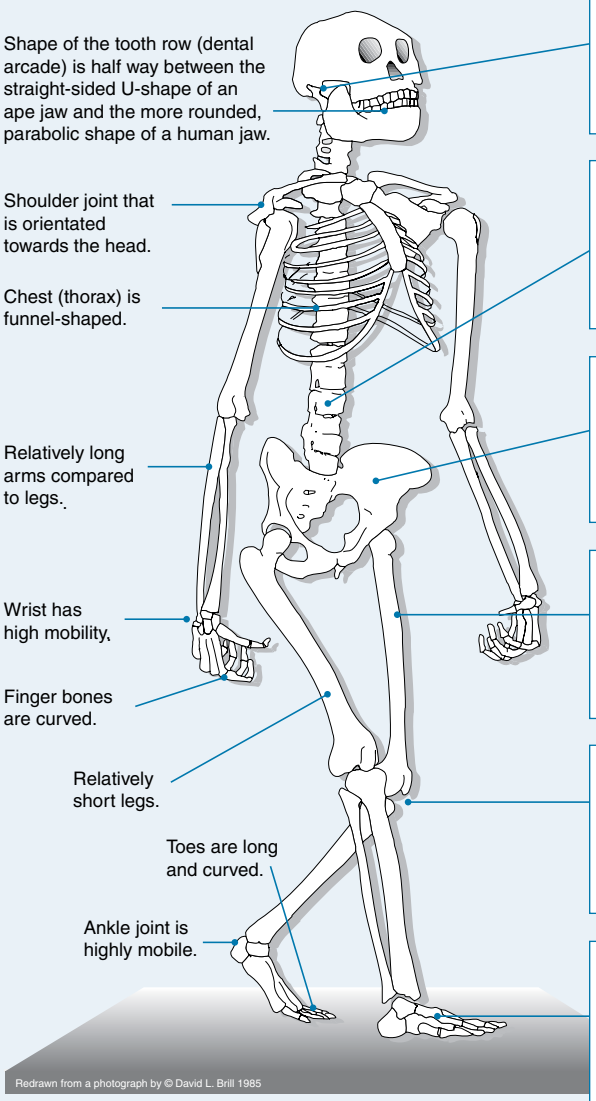
   (a) Lower end of femur: _____

   (b) Foot prints: _____

   (c) Foot skeleton: _____

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Lucy's* ape-like characteristics



*Lucy is the name given to a specimen of ***Australopithecus afarensis***

(a) Position of foramen magnum (where the spine enters the skull):

(b) Spine shape:

(c) Pelvis shape and gluteus muscles:

(d) Femur shape and length:

(e) Knee joint:

(f) Shape of foot and arrangement of toe bones:

2. In the boxes, (a) to (f) above, describe the modifications required for habitual bipedal locomotion (walking).

3. Describe six possible selection pressures that may have encouraged the development of bipedalism in early hominids:

   (a) _____ (d) _____

   (b) _____ (e) _____

   (c) _____ (f) _____

4. Imagine you are on an expedition to a well known hominin fossil site in east Africa. Which part or parts of a hominin fossil skeleton would be most informative in clearly indicating **bipedalism**? Explain your choice:

   _____

5. Describe two selection pressures that may have encouraged the retention of tree-climbing ability in *A. afarensis*:

   (a) _____

   (b) _____

6. Discuss the significance of the **valgus** (carrying) **angle** in hominin bipedal locomotion: _____

   _____

   _____

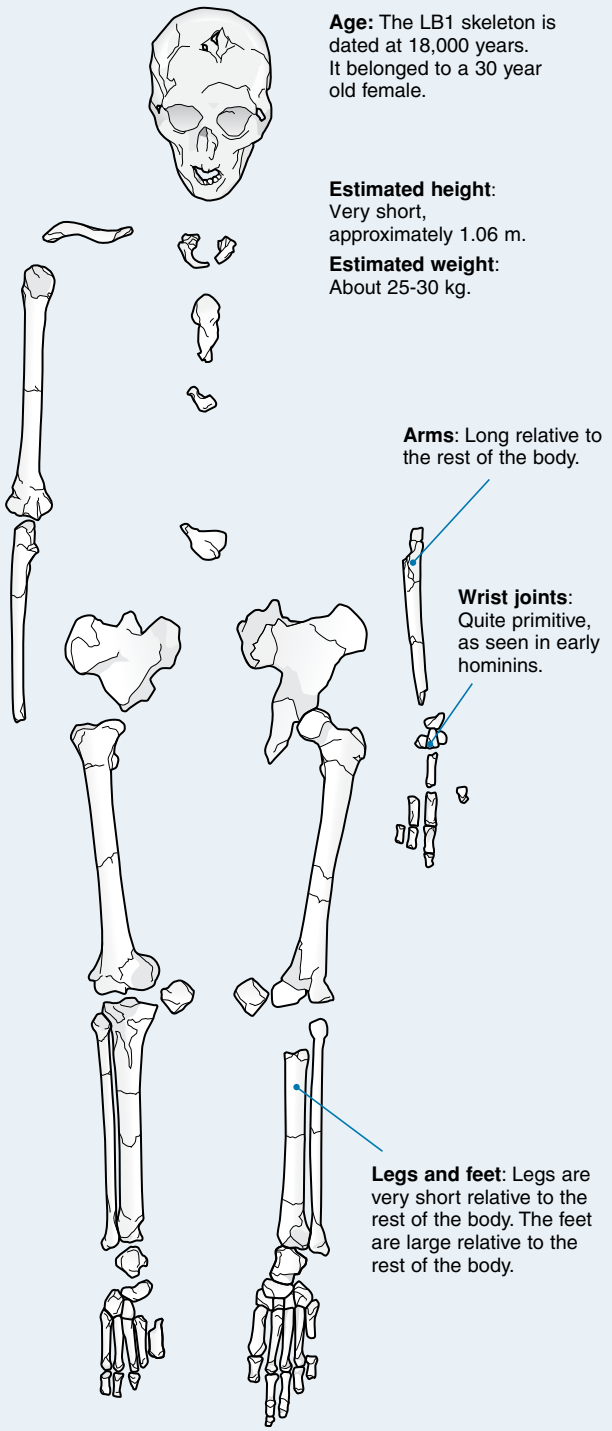
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262 A Controversy in Human Evolution

Key Idea: The discovery of *Homo floresiensis* has completely changed some views about human evolution. It has proved a highly controversial find with several hypotheses provided to explain its position in the evolution of humans.

In 2003, hominin fossils, including an almost complete skeleton, were discovered on the island of Flores, Indonesia. The fossils were astounding for their recent date (18,000 years) and mix of primitive and derived features. This new species, *Homo floresiensis*, was contemporary with moderns, but very small and fully bipedal. Although the brain was very small, its organisation was advanced and stone tools associated with the skeleton indicated well established hunting technology. In contrast to these features, aspects of the skeleton showed primitive features found only in apes and early hominins. *H. floresiensis* probably lived 38,000-13,000 years ago, but the species may date back as far as 95,000 years ago. Its discovery caused widespread controversy and several opposing hypotheses were put forward to explain its place in human evolution. You will analyse the information for these hypotheses in this activity.



Artist's reconstruction of the LB1 skeleton, after William Junger, Stony Brook University



Rosino

Above: The fossils were discovered in Liang Bua, a limestone Cave on Flores Island, Indonesia. The cave contains 12 m of stratified deposits. The remains of modern humans, as well as Homo floresiensis, have been found in the cave.





Avandergeer

A cast of the *Homo floresiensis* fossil LB1 (above, left) compared a microcephalic present day skull (above, right).

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Hypothesis	Key Features	Comments
*Homo floresiensis*is is a new species of *Homo*	The fossils are those of a new hominin species, *H. floresiensis* (*H.f*), and evolved on Flores from an unknown small-bodied and small-brained hominin which had earlier migrated from Africa. This ancestral hominin was perhaps a Dmanisi hominin.	A study of the bones and joints of the arm, shoulder, and lower limbs concluded that *Homo floresiensis*is was more similar to early humans and apes than modern humans. Small brain size coupled with more advanced brain organisation indicate possible parallel evolution of sapiens-like features.
The features of *Homo floresiensis* are the result of island dwarfism	An isolated population of *H. erectus* migrated to Flores and underwent island dwarfism, a phenomenon whereby large animals become smaller in size because of limited resources.	Extinct pygmy elephants on Flores showed this adaptation.
The features of *Homo floresiensis* are the result of disease (pathology)	*H. floresiensis* is a diseased variant of *H. sapiens*. Pathologies proposed to produce the features observed in *H. floresiensis* include: Microcephaly, a neurodevelopmental disorder with various causes, which causes abnormally small brain size	Comparisons of cranial endocasts of 9 microcephalic and 10 normal human brains indicated *Homo floresiensis*is brains to be highly developed in the frontal and temporal regions and bear no resemblance to microcephalic brains. Statistical analysis of skull shapes of healthy modern humans, microcephalic humans, and several ancient human species, as well as *Homo floresiensis*is showed that the three grouped separately, with *Homo floresiensis*is among the ancient humans, providing evidence for it being a separate species.
	Congenital hypothyroidism, which can be caused by factors, including iodine deficiency, which is an endemic public health problem in parts of Indonesia.	Congenital hypothyroidism is endemic in parts of Indonesia but a comparison of the Flores bones with the bones of people affected with hypothyroidism showed no overlap in features. The carpals belonging to LB1 resembled ape carpals and were comparable to carpal bones of *Australopithecus*. It is improbable the shape could be the result of disease.

1. After reviewing the information provided about the *Homo floresiensisis* fossils, choose **one** hypothesis from the table above which you think provides the mostly likely explanation for the physical appearance of *Homo floresiensisis*. Explain why you have chosen your hypothesis, and justify your position using examples.

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263 Trends in Brain Volume

Key Idea: The evolution of a large brain was crucial to our development of language, technology, and culture.

The human brain is responsible for our unique human behavioural qualities. It makes up just 2% of our body weight, but demands about 20% of the body's metabolic energy at rest. This makes the brain an expensive organ to maintain. The selection pressures for increased brain size must have been considerable for additional energy to be made available. The normal human adult brain averages around 1330 cc, but ranges in size between 1000 and 2000 cc. But intelligence is not just a function of **brain size**. There are large mammals, such as elephants and whales, with brain volumes greater than ourselves and yet they are not considered to be as intelligent. It appears that what is more important is the **relative brain size** (brain size relative to body size). Modern humans have a brain volume three times larger than that predicted for an average monkey or ape with our body size. Another important factor is the way in which the brain is organised. Apart from the highly developed cerebrum, two areas of the brain associated with communication have also become highly developed in modern humans: **Broca's area**, concerned with speech, and **Wernicke's area**, concerned with comprehension of language.

Growth in Brain size in Humans and Chimpanzees

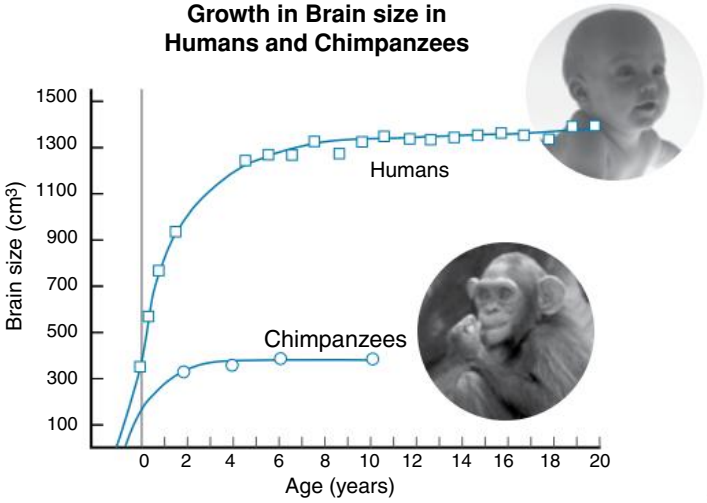


Figure above: In most primates, including chimpanzees, brain growth, relative to body size, slows markedly after birth while body growth continues. In human infants, the slowing of brain growth does not occur until more than a year after birth, which results in larger brain masses for humans than for chimpanzees at any given age (or body weight).

Brain Volume for Hominin Species

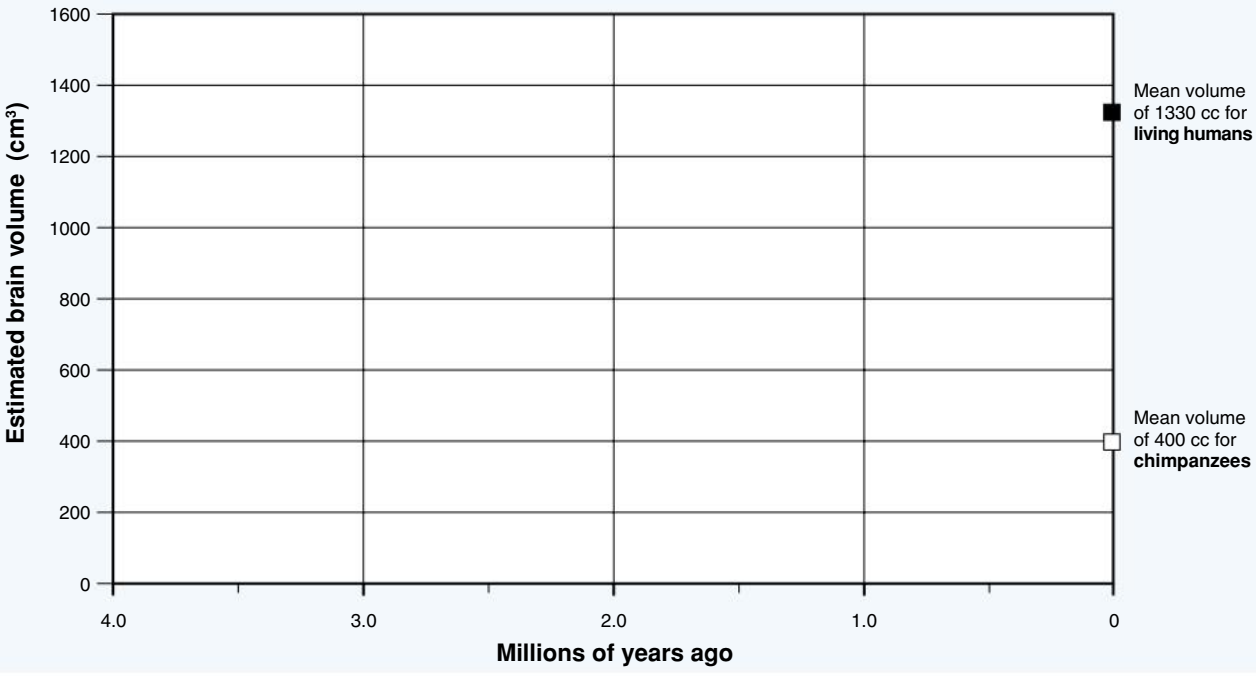
Hominin species	Years ago (mya)	Average brain Volume (cm³)
Australopithecus afarensis	3.5	440
Australopithecus africanus	2.5	450
Paranthropus robustus	2.0	520
Paranthropus boisei	1.5	515
Homo rudolfensis	2.0	700
Homo habilis	1.8	575
Homo ergaster	1.8	800
Homo erectus	0.5	1100
Homo heidelbergensis	0.2	1250
Homo neanderthalensis	0.05	1550
Homo floresiensis	0.095*	380
Early *Homo sapiens*	0.08	1450

* *H. floresiensis* may have lived as recently as 13,000 ya

Table above: A generalised summary of the changes in estimated brain volume recorded from the fossil remains of hominins. The dates for each species are generally the middle of their time range for long-lived species or at the beginning of their time range for short-lived species.

1. Plot the data in the table on the estimated *Brain Volume for Hominin Species* (above) onto the graph below.

Changes in hominin brain volume over time

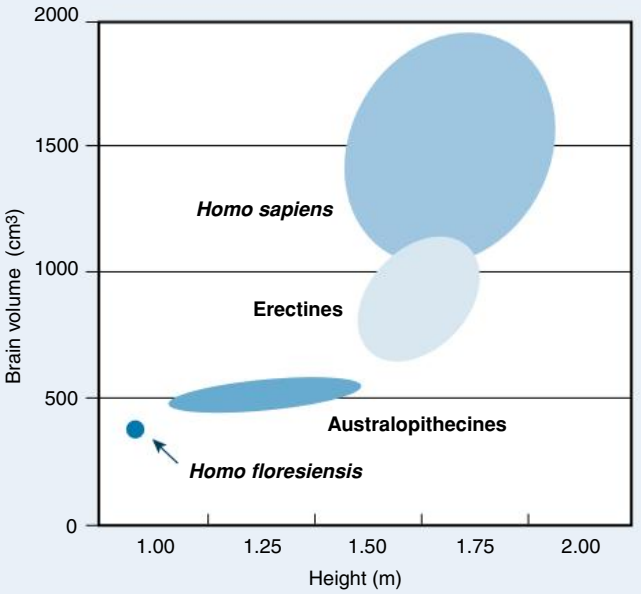


2. There were two 'bursts' (sudden increases) of brain expansion during human evolution. **Indicate on the graph** you have plotted where you think these two events occurred.

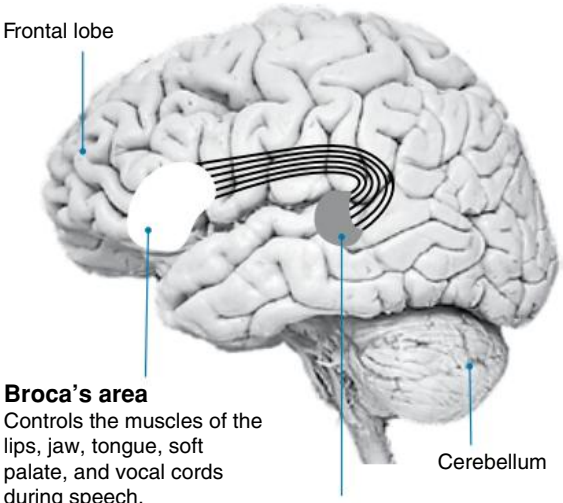
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Brain Size vs Body Height in Hominins



Brain size can be correlated with body height in hominins. Three distinct clusters emerge, indicating three phases of evolutionary development. *Homo floresiensis*, found on the Indonesian island of Flores, clearly falls outside these clusters. Its brain size to body size ratio is similar to that of the *Australopithecines*, but key aspects of its morphology, such as its small canine teeth and organisation of the brain, identify it as *Homo*. In addition, the Flores finds were associated with relatively advanced stone tools.



Modern Human Brain

The human brain is very large for a primate of our size, but this may not be as important as its internal organisation. The most important specialisation of the human brain is the capacity for language: a result of the development of **Wernicke's** and **Broca's areas**. Specific differences associated with the left and right hemispheres of the brain are associated with these specialisations.

3. Why is brain volume alone not a reliable indicator of intelligence? _____

4. Explain the significance of the high energy requirement of a relatively large brain: _____

5. Comment on the significance of the brain/body size growth curve in humans compared with other primates:

6. (a) With respect to stature and brain size, comment on the position of *Homo floresiensis* with respect to other hominins:

   (b) Comment on the significance of the Flores finds: _____

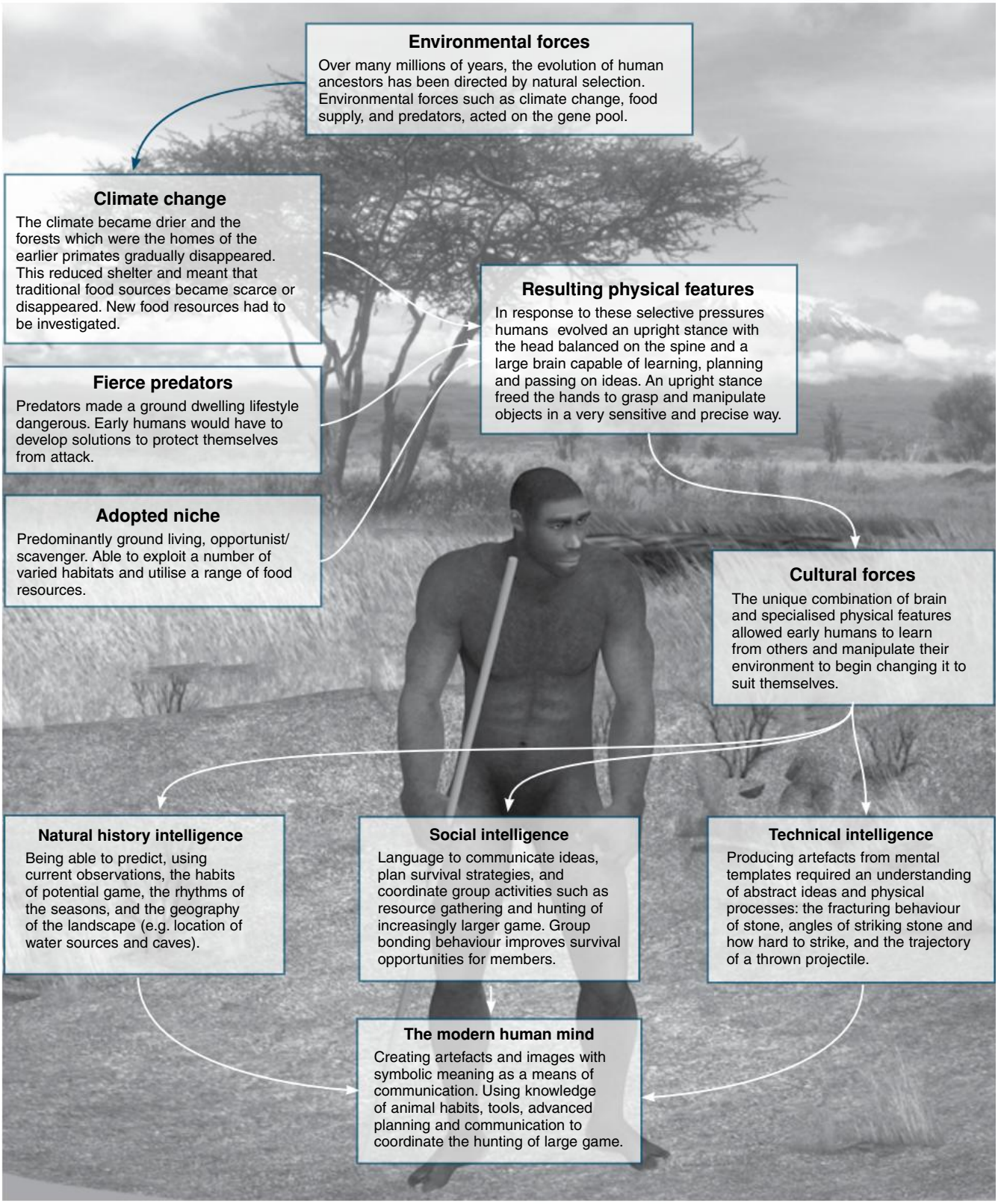
7. Describe a likely **selection pressure** for brain development in early humans: _____

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264 Cultural Evolution

Key Idea: Cultural evolution is a term used to describe the transmission of and changes to knowledge and ideas over successive generations.

In addition to the physical evolution of humans, ideas and behaviours also evolved as they were learned and passed on to offspring. This non-genetic means of adaptation, called **cultural evolution**, further enhanced the success of early humans.



1. Explain what is meant by **cultural evolution**: _____

_____

_____

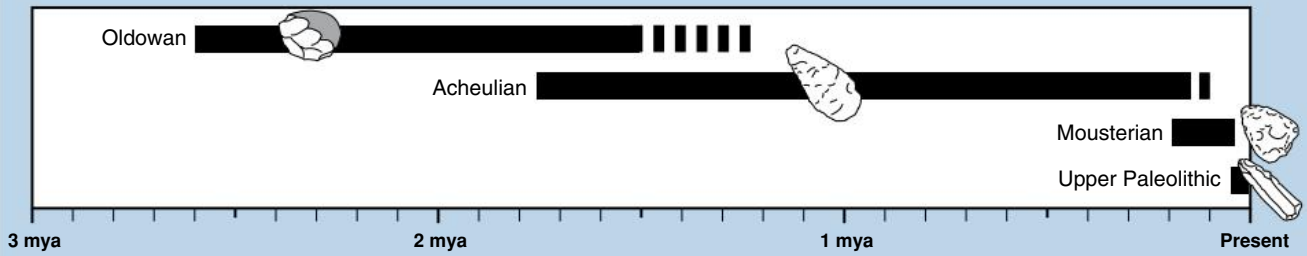
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265 Palaeolithic Tool Cultures

Key Idea: The development of stone tools is a defining characteristic of *Homo*. Specific types can be matched specific time periods and species.

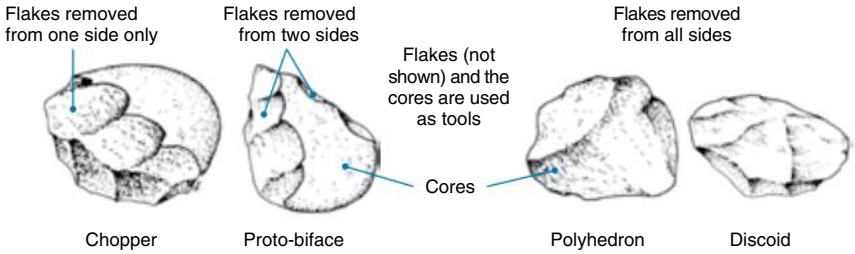
The **Palaeolithic** (Old Stone Age) is a period of early cultural development spanning the emergence of the first stone tools about 2.6 mya in eastern Africa, until the development of sophisticated tool kits in the Mesolithic (Middle Stone Age) about 10,000 ya. These tool cultures are known mostly by their stone implements. While other materials, such as wood, were probably also used, they did not preserve well.

Timeline of Stone Tool Technologies



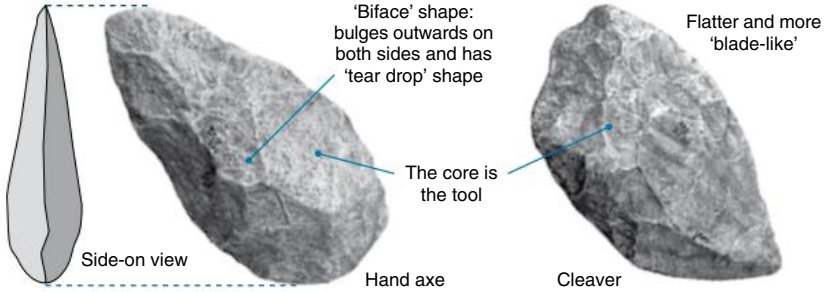
Oldowan (pebble) tool culture

Probably made by *Homo habilis*, these tools were simple river-worn pebbles that were crudely fashioned with a minimum of flakes being removed. These tools typically had flakes knocked from several angles to produce a core with a cutting edge (e.g. chopper, discoid, polyhedron). Although the cores may have been used as tools, it is known that the sharp flakes were also useful in cutting.



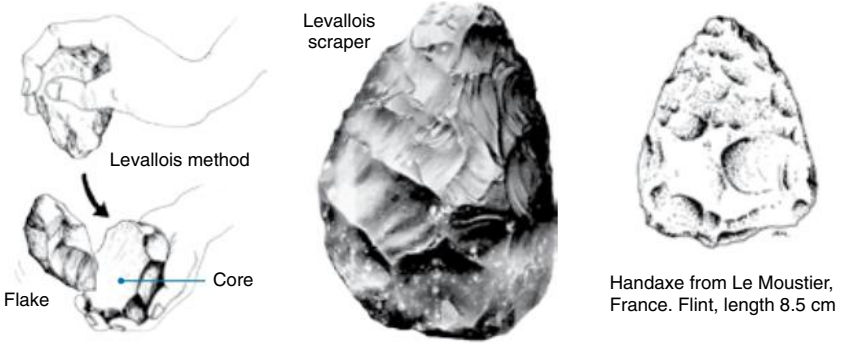
Acheulian tool culture

The product of *Homo erectus* and archaic *Homo sapiens*, these tools were typically 'tear drop' in shape and were carefully crafted with a slight bulge on each broad surface (called a bi-face). They ranged greatly in their size and are often referred to as hand axes although it is not clearly understood how they were used. They differ markedly from the earlier pebble tools in that there appears to be a standard design and each tool is manufactured using a great many more blows to remove flakes.



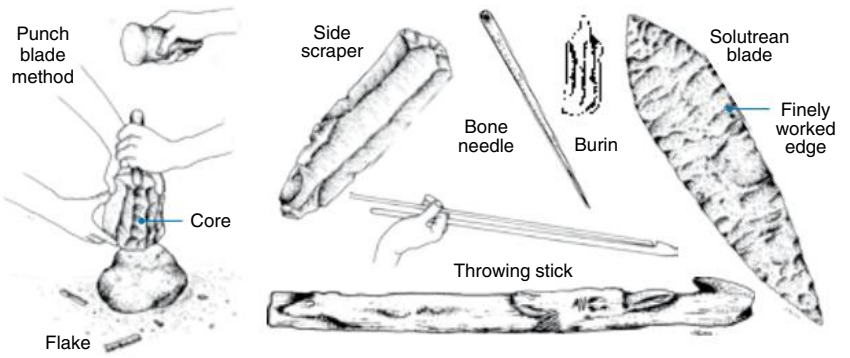
Mousterian tool culture

The **Neanderthals** developed a more refined tool culture than the earlier Acheulian. Flint finally became a sought after material to produce stone tools. The advantage of this rock was the very predictable way in which it would chip when struck with another hard object. Much finer workmanship was possible. A particular tool making technique from this period is known as the **Levallois method**. It involves the preparation of a core and striking off a large oval flake which is then retouched on one surface only (see the photograph on the right; the retouched surface is visible).

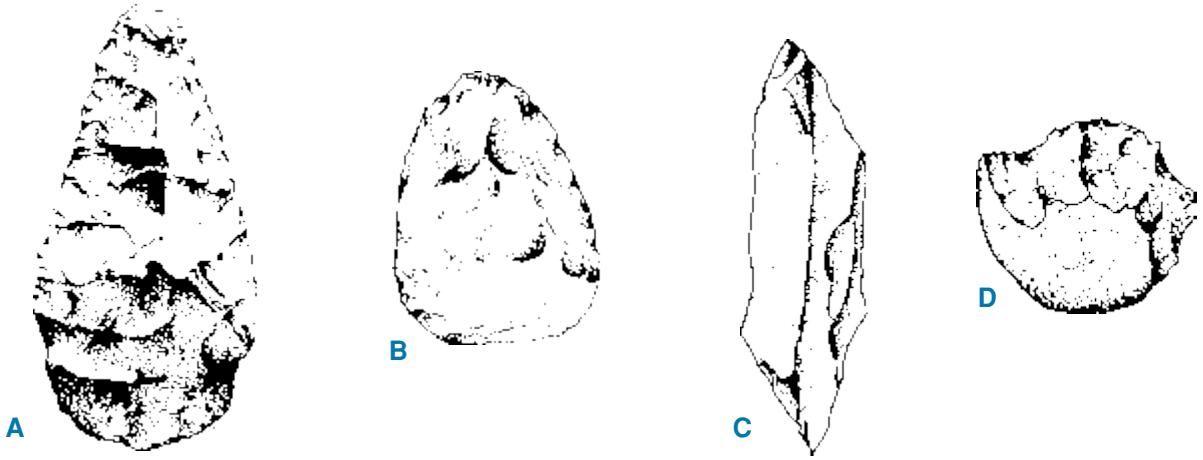


Upper Palaeolithic tool culture

There was a rather sudden increase in the sophistication of tool making about 35,000 to 40,000 years ago. Both the **modern** *Homo sapiens* and the last of the Neanderthals produced flint tools of much finer workmanship using a technique called **punch blade**. Long, thin flakes are removed and shaped into a large number of different tool types. European sub-cultures (traditions) include the **Solutrean**, **Magdelanian**, and **Aurignacian**. Other material such as bone, ivory and antler became increasingly utilised to produce very fine tools such as needles.



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1. Name the culture associated with each of the tools above (A-D) and describe the features that help identify them:

   (a) Tool **A** culture: _____

   _____

   (b) Tool **B** culture: _____

   _____

   (c) Tool **C** culture: _____

   _____

   (d) Tool **D** culture: _____

   _____

2. Identify the **hominin species** associated with, and the approximate time period for, each of the tool cultures below:

   (a) Oldowan: _____

   (b) Acheulian: _____

   (c) Mousterian: _____

   (d) Upper Palaeolithic: _____

3. Describe the general trends in the design of the stone tool from **Oldowan** to **Upper Palaeolithic** cultures:

   _____

   _____

4. The tools that are recovered from early human prehistoric sites are almost invariably stone, bone or ivory. Explain why tools made from other materials are almost never recovered from these sites:

   _____

   _____

5. Name the materials used to make tools in the **Upper Palaeolithic** culture that were seldom used in earlier cultures:

   _____

6. Explain why the makers of the **Oldowan** tools were not able to produce designs that were as sophisticated as those of the Upper Palaeolithic:

   _____

   _____

   _____

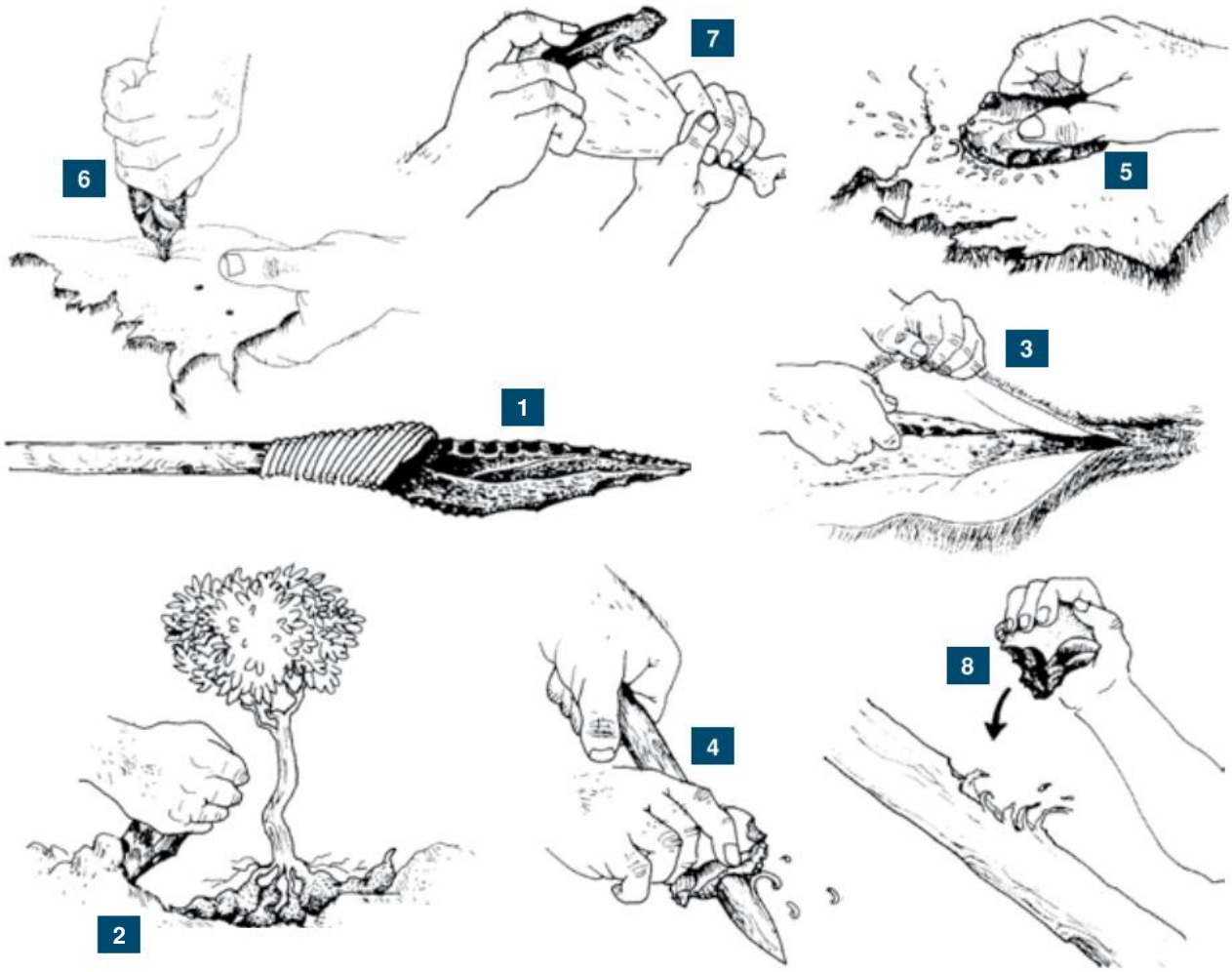
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266 Palaeolithic Tool Use

Key Idea: Stone tool technology can be adapted for numerous purposes including spear heads, blades, and axes.

It is possible to infer how the stone tools of early hominins were used by studying how similar tools have been used by recent 'stone age' societies. People using only stone-based technology were still in existence well into the first half of last century. Hunter-gatherer people existed in places like the Kalahari desert in south west Africa, the Australian outback, and some of the more remote areas of South East Asia and South America. Anthropologists studying these primitive cultures gathered valuable insights into how our ancestors may have lived.



1. Match each of the diagrams above with the **description** of their function in the table below (place 1-8 in number column):

	No.	Name of Tool	Description
(a)			This tool was probably used to scrape the fat and sinew from the underside of a freshly killed animal, in preparation for curing.
(b)			The curved, sharp edge of this tool would probably have been used to shave wood chips from spear heads.
(c)			Later forms of this tool were probably used to skin and dismember carcasses.
(d)			A simple tool used as an axe, probably to cut into wood or possibly to dismember a carcass.
(e)			Early forms of this tool may have been used as a pick-like tool to expose root tubers growing under plants.
(f)			Used as a knife, this tool had only one side with a sharp cutting edge so that pressure could be applied to the blunt edge.
(g)			Hafted to a pole with greased sinew or plant fibre, this flint tool would have provided an effective cutting edge for the spear.
(h)			The sharp narrow point of this tool makes it an effective drill when twisted back and forth. In this way, holes could be made in materials such as hides, wood and ivory.

2. Assign each tool with their **correct** name from the list below and place the correct name in the table above:
Side scraper, borer, denticulate tool, spear point, chopper, backed flake, early hand-axe, late hand-axe

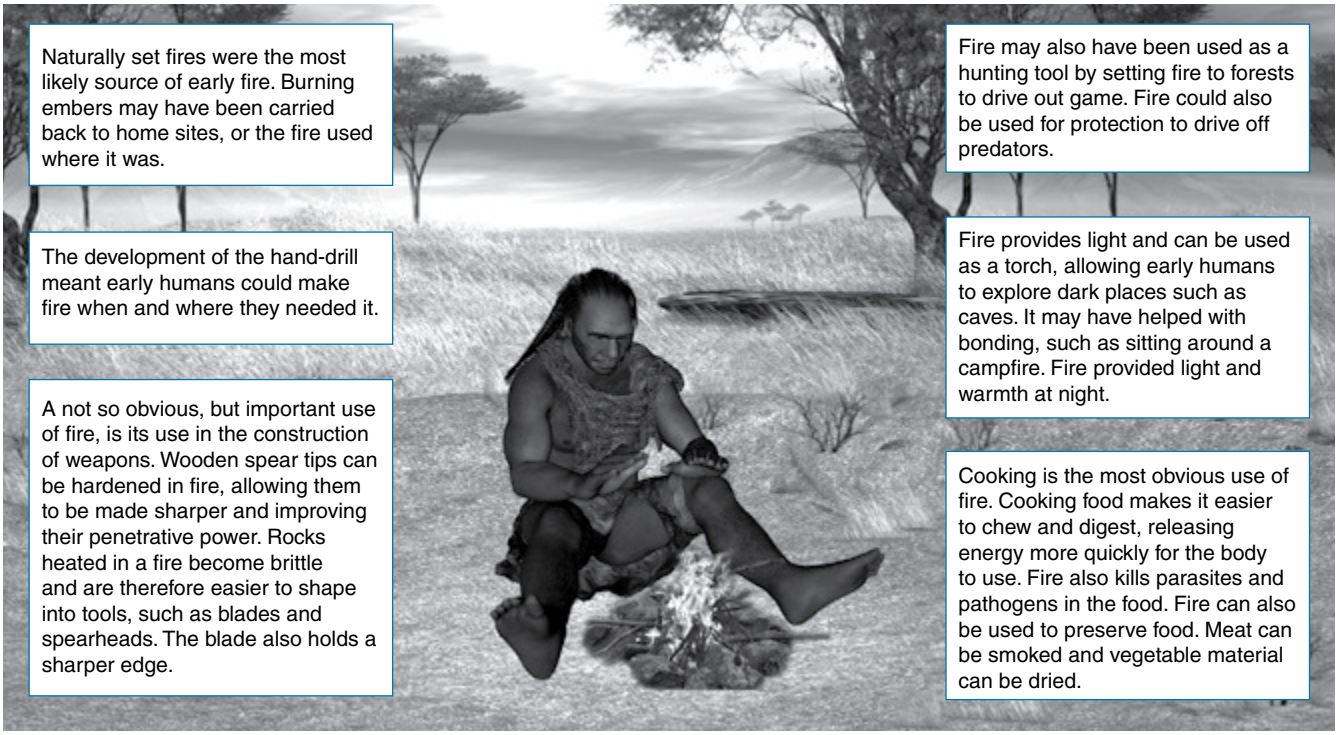
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LINK 265 KNOW

267 Fire

Key Idea: The controlled used of fire by early humans allowed them to eat a greater range of foods and to expand their range into darker and colder places.

Just when and how early human ancestors began using fire is debated and may never be precisely known. It is likely that early fire was "captured" from the wild, e.g. from fires started by natural events such as lightning. Whatever its origin, the controlled use of fire changed the course of both physical and cultural evolution. Possible evidence exists for the use of fire up to a million years ago, but real evidence of controlled use of fire dates back only a few hundred thousand years.



Naturally set fires were the most likely source of early fire. Burning embers may have been carried back to home sites, or the fire used where it was.

The development of the hand-drill meant early humans could make fire when and where they needed it.

A not so obvious, but important use of fire, is its use in the construction of weapons. Wooden spear tips can be hardened in fire, allowing them to be made sharper and improving their penetrative power. Rocks heated in a fire become brittle and are therefore easier to shape into tools, such as blades and spearheads. The blade also holds a sharper edge.

Fire may also have been used as a hunting tool by setting fire to forests to drive out game. Fire could also be used for protection to drive off predators.

Fire provides light and can be used as a torch, allowing early humans to explore dark places such as caves. It may have helped with bonding, such as sitting around a campfire. Fire provided light and warmth at night.

Cooking is the most obvious use of fire. Cooking food makes it easier to chew and digest, releasing energy more quickly for the body to use. Fire also kills parasites and pathogens in the food. Fire can also be used to preserve food. Meat can be smoked and vegetable material can be dried.

1. List four uses of fire: _____

2. (a) Describe two important consequences of cooking food: _____

   (b) Explain how cooking food provided a selection pressure for a smaller jaw and teeth: _____

3. Discuss the influence of fire on human cultural evolution: _____

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268 Shelter and Clothing

Key Idea: The development of clothing and techniques to build shelters allowed the expansion of humans into colder climatic areas.

Human ancestors would have had a far greater amount of body hair than humans today. The move into hot open grassland provided a selection pressure for the reduction of body hair. However this then limited the ability to survive in cold climates as humanity migrated north out of Africa into Europe and beyond. The use of animal skins or woven plant material to make clothing provided a way of keeping warm. Building shelters allowed humans to shelter from both the weather and possible predators.

Early humans and their ancestors are often depicted in popular texts wearing animal skins. However there is little direct evidence of humans wearing clothes until 26,000 years ago (the evidence is a finely worked bone needle). Evidence from burial sites also shows humans were adorning themselves (or at least their dead) with necklaces as far back as 35,000 years ago. Evidence from the evolution of body lice suggests human ancestors began wearing clothes some 100,000 years ago.



This finely worked bone needle is almost identical in shape to needles used today. This needle is dated between 17,000 and 10,000 years old.

Didier Descouens CC 3.0



Hide drying on rack

Although often called cavemen, human ancestors probably didn't start using caves regularly as shelter until they could control the use of fire. Bones of early australopithecines or *Homo* found in caves were likely taken there by predators or been washed in by ancient streams. Some of the first evidence of caves being used by humans is from the Atapuerca cave in Spain. It is dated at 1.2 million years old, but it is difficult to tell the exact relationship of the fossils to the cave.





The first evidence of humans building purpose-built shelters dates to around 400,000 years ago. Post holes in the ground indicate poles were used as scaffolding for the shelter. Preserved bones from around 17,000 years ago in the Ukraine show humans there used mammoth bones and tusks as scaffolding, probably because there would have been few trees available to make wood scaffolding from.

1. (a) Identify the evidence for humans building purpose built shelters: _____

   _____

   (b) Identify the evidence for humans making clothing: _____

   _____

   (c) Suggest a reason why there is so little evidence of humans building shelters or making clothes: _____

   _____

   _____

2. Suggest a reason why caves where probably not regularly used by humans and there ancestors until the controlled use of fire was developed:

   _____

   _____

   _____

3. What is the main purpose of clothing? _____

   _____

   _____

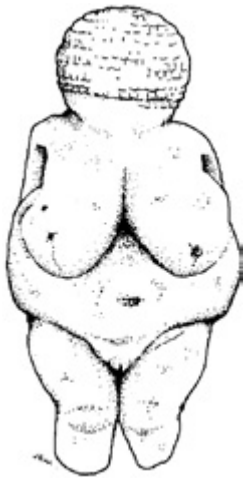
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LINK 269 LINK 267 LINK 265 LINK 264 KNOW

269 Art and Spirituality

Key Idea: A cultural explosion took place in the form of prehistoric art and new kinds of tools attributed to modern humans about 35,000 years ago.

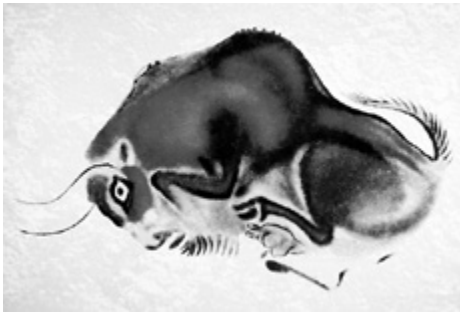
Until recently, it was believed that art and spiritual beliefs first developed with the arrival of modern humans, particularly in Europe. The beginning of a period 35,000 years ago marks a dramatic cultural development occurring simultaneously over large parts of western and eastern Europe. Growing evidence suggests Neanderthals too were culturally sophisticated. Not only did they bury their dead, but they appear to have adorned their bodies, used necklaces, and made and used flutes. The stimulus for the new cultural development, was probably a need to represent ideas about the unknown, such as death, hunting success, and fertility, in a concrete way. A wide range of materials were used to do this. Ivory, bone, clay, and stone were used to create sculptures, and the walls of rock shelters and caves were adorned with drawings, paintings, and bass-relief (sculptures that stand out from the rock wall).



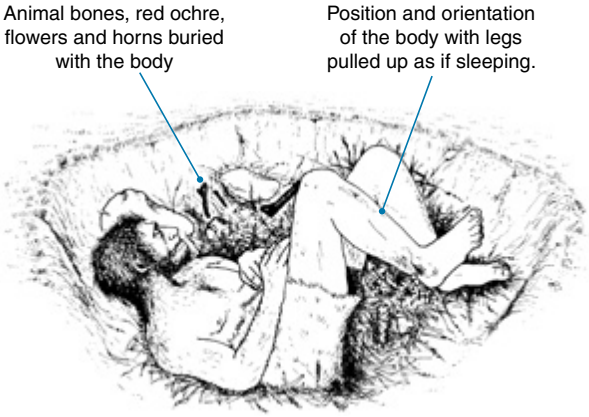
Sculpture: One of the earliest prehistoric works of art is the Venus of Willendorf (left), from Austria (10 cm high). The small figurine, carved from limestone about 30,000 years ago, has exaggerated breasts, buttocks, and body fat; these were thought to be desirable traits among women to enhance fertility and survival in periods of deprivation.



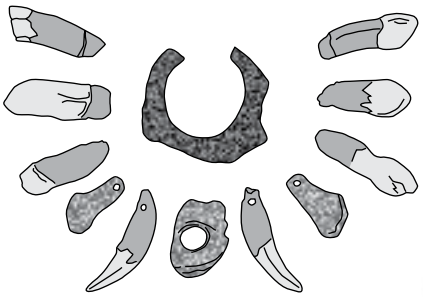
Music: Made of bird-bone, this 25,000 year old flute (above) is one of the earliest instruments providing evidence of music. A similar flutelike piece of cave bear bone has been found at a Neanderthal hunting camp in Slovenia. The bone, dated at between 43,000 and 82,000 years ago, suggests that Neanderthals may have made music.



Rock art: Many examples of rock art are found in southern France and northern Spain. However, rock art was not restricted to Europe; the earliest examples may come from Australia. The pigments (charcoal, manganese oxide, and ochre) were applied to grease smeared on the rock surface.



Neanderthal burials: The Neanderthals of Europe and Southwest Asia buried their dead in a way that showed signs of ritualisation. The grave is usually characterised by certain items found buried with the body. The position and orientation of the body are consistently the same. Some experts have questioned the validity of these so-called burials and claim the finds are the result of coincidences and later disturbances.



Neanderthal necklace: The necklace above was recovered from a late Neanderthal site. Was the necklace made by the Neanderthal, or was it traded or stolen from modern humans? Whatever the answer, it suggests that Neanderthals appreciated such objects.



Magic and ritual: Painted about 15,000 years ago on a cave wall at Trois Féres, in the Pyrenees Mountains, this creature (above right) may have been a sorcerer dressed in animal skins. Rituals involving spiritual re-enactment may have been conducted to ensure successful hunting.

1. (a) Identify three items that may have been found with the body in a Neanderthal grave: _____

_____

(b) Describe the usual position and orientation of skeletons found at Neanderthal burial sites: _____

_____

_____

_____

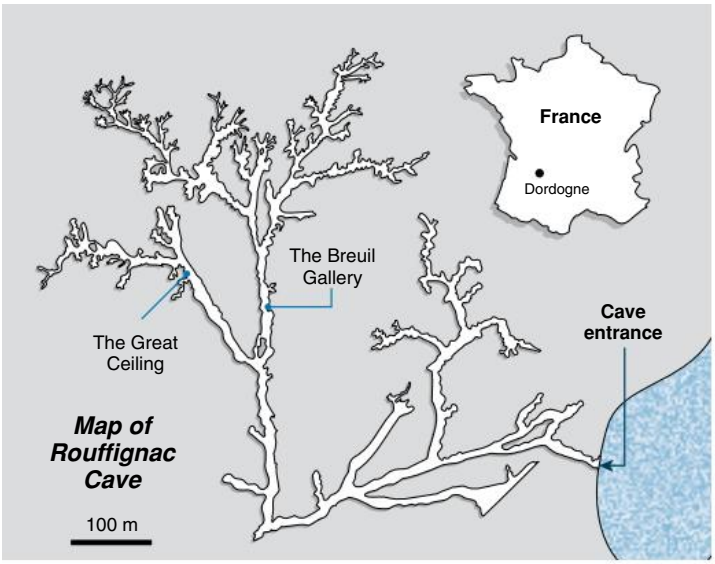
2. Describe two recent findings that support a view that Neanderthals were more sophisticated than previously thought:

(a) _____

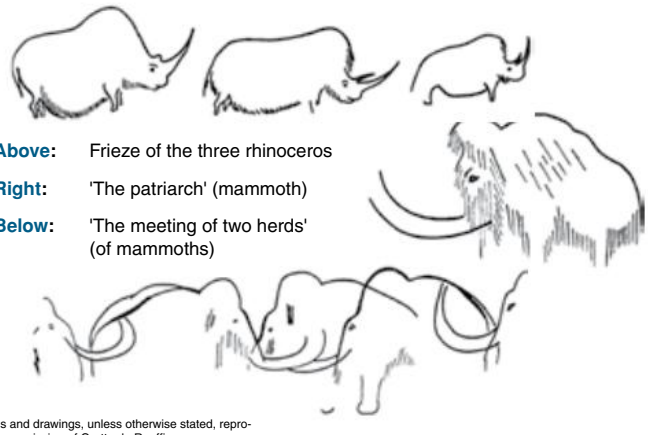
(b) _____

KNOW LINK 255 LINK 264

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Rouffignac cave, in the Dordogne Valley in southwestern France, is a huge cavern with 10 km of galleries. It is remarkable for its large number of prehistoric paintings dating from the mid to late Magdelanian Period (~11 000 BC). The first drawings are about 750 m from the entrance and continue deep underground, to the end of the galleries. It is interesting to note that over 150 paintings of the mammoth in this cave represent half the known paintings of this animal. Early human inhabitants must have competed with cave bears for cave occupancy. Early humans often created their paintings in the most remote and inaccessible parts of the cave; an argument used in favour of a religion centred on the hidden forces of the Earth. Below and right are some typical examples of these prehistoric drawings. Earlier rock engravings have been dated (disputed by some) in Australia at >60,000 years; this is more than twice the age of the oldest European art.



Above: Frieze of the three rhinoceros

Right: 'The patriarch' (mammoth)

Below: 'The meeting of two herds' (of mammoths)

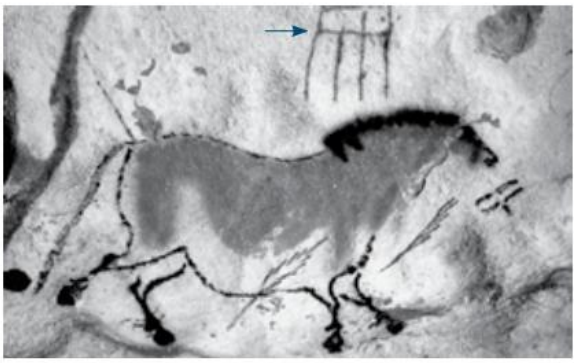
All photos and drawings, unless otherwise stated, reproduced by permission of Grotte de Rouffignac



Detail of the head of a rhinoceros from the Great Frise in the **Breuil Gallery**, Rouffignac Cave. Like the mammoth, the woolly rhinoceros was common to Europe before becoming extinct 12,000 years ago.



The **Great Ceiling** is the heart of the sanctuary at Rouffignac. The collection of drawings depicts a mammoth and ibex. It was common for drawings to be made over the top of one another.



Corel

This painting of a horse is one of many images painted on the ceiling of the famous Lascaux cave in the Dordogne, France. Note the abstract markings (arrowed) for which there is no explanation.

3. Describe the relationship the early humans had with the mammoth and woolly rhinoceros: _____

4. Describe a probable purpose for the drawings and paintings appearing in the cave art: _____

5. Describe the likely function of the 'Venus' statues in Palaeolithic life (see the *Venus of Willendorf* on the previous page):

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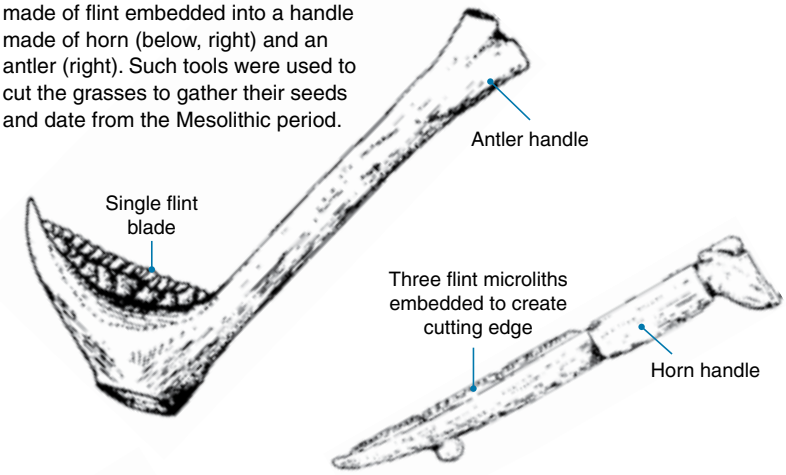
270 Mesolithic Culture

Key Idea: The Mesolithic period is marked climatically by the change from glacial to warmer climates and archaeologically by the refinement and further development of human tools.

The **Mesolithic** (Middle Stone Age) period occurred in Europe 12,000-3000 years ago, as the last glacial period ended. The tools produced at the time were small bladed geometric stone tools (**microliths**), and were often fitted into a handle of wood or bone. Mesolithic people used a wide variety of hunting, fishing, and food gathering techniques, which may have arisen because the warming climate would have resulted in increased productivity. The Mesolithic occurred during the current Holocene epoch, which is relatively warm compared to the previous epoch, the Pleistocene. The warmer climate resulted in glacial retreats, the growth of forests in Europe and deserts in North Africa, and the disappearance of the animals hunted during the glacial period.

Tools of the Mesolithic Period

Sickles: These two sickles are made of flint embedded into a handle made of horn (below, right) and an antler (right). Such tools were used to cut the grasses to gather their seeds and date from the Mesolithic period.





Bone fish hook: This fish hook dates from the Mesolithic period and was found in Sweden.



Didier Descouens

Harpoon: This flat harpoon is made of bone, and dates between 12,000 and 9500 years ago. It was found in France.



Didier Descouens

Microlith: Made of flint or chert (a sedimentary rock). Microliths formed the points of hunting weapons such as spears. This microlith was found in the Tourasse cave, France.

1. The **Mesolithic culture** replaced the Upper Palaeolithic culture. When did the Mesolithic culture begin?

2. (a) Describe the key features that characterise the Mesolithic culture:

   (b) How did the Mesolithic culture differ from the Upper Palaeolithic culture?

3. Explain the significance of the warmer climate experienced during the Mesolithic period:

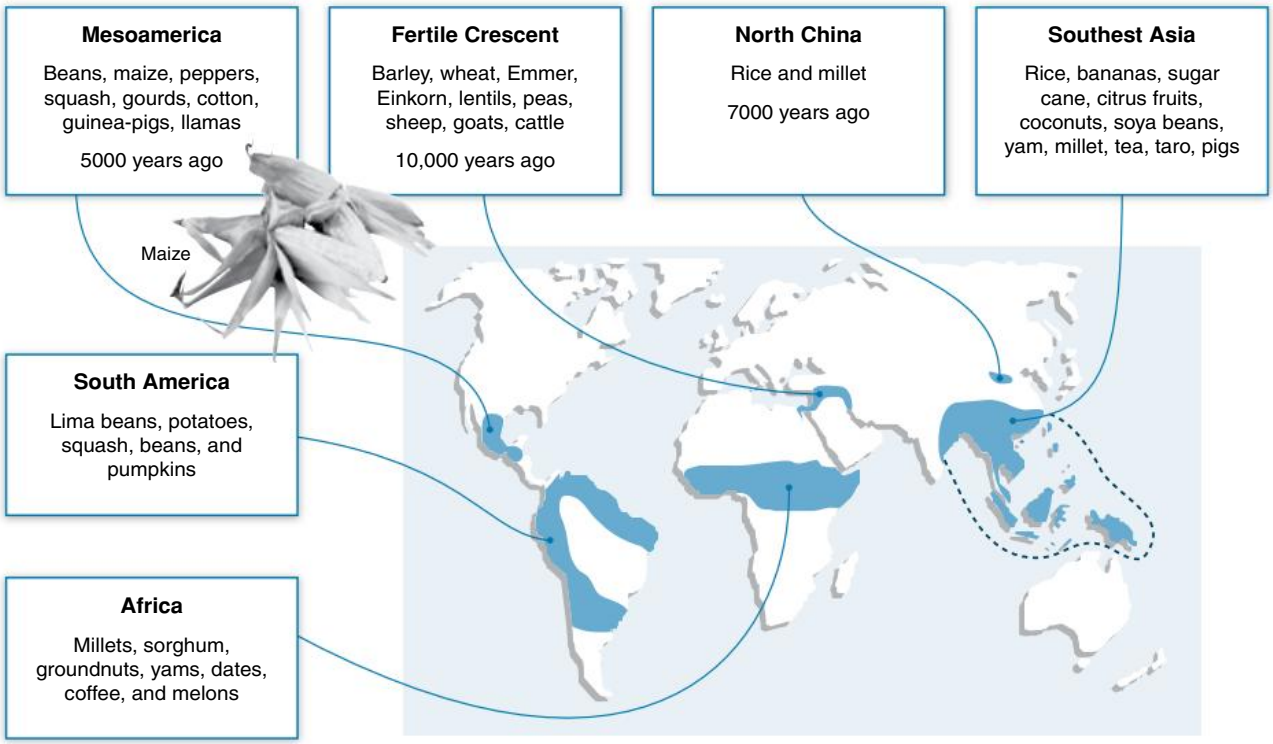
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271 Neolithic Culture

Key Idea: The Neolithic is characterised by advances in farming and animal domestication practices, the development of crafts (e.g. pottery and weaving), the use of polished stone and flint tools, and the development of permanent settlements.

The date of the **Neolithic** (New Stone Age) culture varies with geographic location. In the middle East it dates from about 10,000 years ago, and in Europe from 4000-2000 years ago. Domestication of animals and the development of farming allowed a shift away from the hunter-gatherer economy of the Mesolithic to a food producing culture. Not all individuals had to be involved in food gathering activities, and some people developed specialised craft skills (e.g. potters). As permanent settlements developed, ideas and knowledge could be more easily transferred between people, resulting in a rapid expansion of cultural evolution during this period.

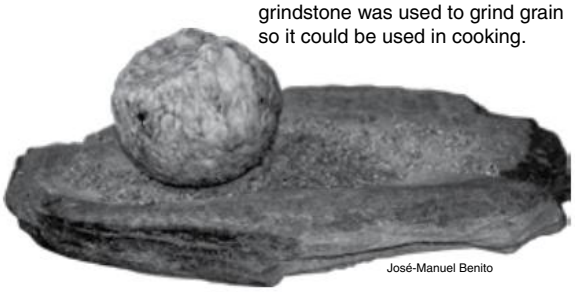
The Origin of Agriculture



Domesticated animal	Wild ancestor	Region of origin	Date (years ago)
Dog	Wolf	many places?	13,000
Goat	Bezoar goat	Iraq	10,000
Sheep	Asiatic mouflon	Iran, Iraq, Levant	10,000
Cattle	Aurochs	Southwest Asia	10,000
Pig	Boar	Anatolia	9000
Domestic fowl	Red jungle fowl	Indus Valley	4000
Horse	Wild horse	Southern Ukraine	6000
Arabian camel	Wild camel	Southern Arabia	5000
Bactrian camel	Wild camel	Iran	4500
Llama	Guanaco	Andean plateau	6000
Water buffalo	Indian wild buffalo	Indus Valley	4500
Ass	Wild ass	Northeast Africa	5500

Each domesticated animal was bred from the wild ancestor. The date indicates the earliest record of the domesticated form.

Grindstone: This Neolithic grindstone was used to grind grain so it could be used in cooking.



José-Manuel Benito



Sandstein

Neolithic people produced a wide range of purpose-specific tools to harvest, store, and prepare food. The photo, above, displays food and cooking items retrieved from a Neolithic site in Europe. The containers are made of antlers and wood.

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LINK 275 LINK 273 LINK 264 KNOW

Dr J.F Burka: http://en.wikipedia.org/wiki/File:Orkney_Skara_Brae.jpg

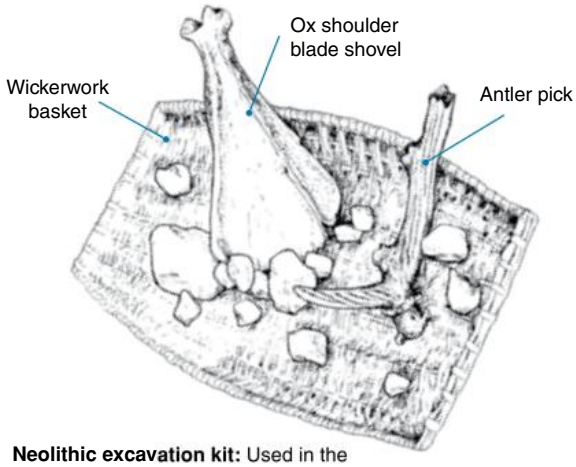
The Development of Permanent Settlements



Neolithic village at Skara Brae, Scotland.

The development of agricultural practices significantly changed the lifestyle of Neolithic people. The hunter-gatherer lifestyle gave way to food production using agricultural techniques. The need to tend to crops and animals resulted in a shift away from a nomadic lifestyle to living in permanent settlements.

Skara Brae, Scotland, is the location of Europe's most complete stone-built Neolithic village (above). It consisted of ten houses, and was occupied between 3180 and 2500 years ago. The people of Skara Brae raised cattle and sheep, and may have cultivated barley. The presence of fish bones and shells amongst the ruins suggest their diet was supplemented with seafood from the nearby ocean.



Neolithic excavation kit: Used in the construction of massive earthworks in Britain (such as Avebury Stone Circle). The pick made from an antler was used to loosen chalk, while the ox shoulder blade was used to shovel the soft rock into a wickerwork basket.



Arrow head: Made of flint, this finely worked Neolithic arrow head was recovered from the Algerian Sahara desert and is about 6000 years old.

1. The **Neolithic culture** replaced the Mesolithic culture. When did the Neolithic culture begin

   (a) In the middle East? _____

   (b) In Europe? _____

2. Describe the key cultural developments that characterise the Neolithic culture: _____

3. Why was the shift of Neolithic people from hunter-gatherers to farmers an important step in cultural evolution?

4. How did the establishment of permanent settlements influence cultural evolution during the Neolithic period?

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272 KEY TERMS: Did You Get It?

1. INSTRUCTIONS: Test your vocabulary for this and the previous chapter by matching each term to its correct definition.

Acheulian tools

bipedalism

Broca's area

cultural evolution

Mousterian tools

multiregional hypothesis

Oldowan tools

replacement hypothesis

valgus angle

Wernicke's area

A Area of the brain that allows the processing and comprehension of speech patterns.

B Hypothesis that modern humans evolved from several different groups of *Homo erectus* in several regions of the world, with gene flow occurring between regional groups.

C A tool type sometimes referred to as pebble tools and associated with *Homo habilis*.

D Tear drop shaped stone tools associated with *Homo erectus* and archaic *Homo sapiens*.

E Hypothesis that states that humans moved out of Africa and displaced other Homo species living in the rest of the world. Also called *Out of Africa*.

F The habit and action of walking upright on two feet.

G Area of the brain that allows for the production of speech sounds by controlling the jaw, tongue, lips, and vocal cords.

H The change over time in accumulation of knowledge, rules, standards, skills, and mental abilities that humans use to survive.

I Advanced tool type associated with Neanderthals. Made by striking a flake and retouching one side of it (the Levallois method).

J The carrying angle of the femur to the knee, which in humans and their ancestors brings the knee under the centre of gravity.

2. (a) Identify the type of tool labelled A (right): _____

(b) What features allowed you to identify the tool? _____

_____

_____

_____

(c) During what time span was this tool used? _____

_____



3. Compare and contrast the jawbones labelled B and C (right). What features identify the jaw labelled B as being of ancient human ancestry and the jaw labelled C as being from modern humans?

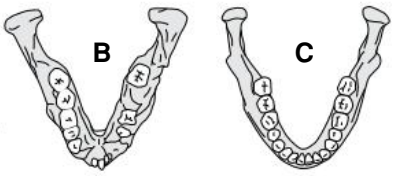
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4. The drawing labelled D is a modern human skull. What features identify it as such?

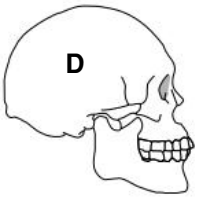
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QLD As required
VCE Unit 4 (2)
WA ATAR Unit 3
SA Theme M, O

Human Intervention in Evolution

Key terms

ancestor
DNA profiling
embryonic stem cell
exogenous
gene cloning
gene therapy
genetic counselling
genetic screening
inbreeding
line-breeding
out-crossing
polyploidy
selection pressure
selective breeding (= artificial selection)
stem cell
therapeutic cloning
transformation
vector

Selective breeding

Learning aims and skills

Activity number

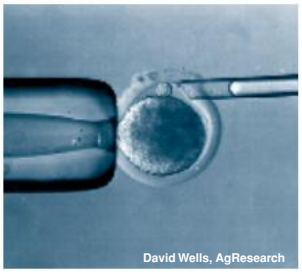
☐ 1 Describe how humans use selective breeding (also called artificial selection) to select for desirable traits while selecting against undesirable traits (thereby affecting and limiting the gene pool of specific organisms). Describe examples to illustrate your understanding of how it operates. If required, compare and contrast selective breeding and natural selection. 273-276

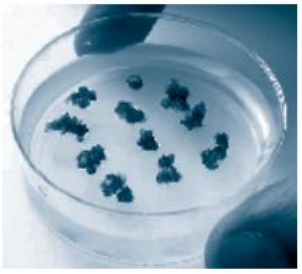
☐ 2 Describe how selective breeding selection has created phenotypic and genotypic change in populations. Examples include:

(a) The development of modern domestic breeds from their wild ancestors. Recognise the role of inbreeding, line-breeding, and out-crossing in "fixing" desirable traits. 273 274

(b) The development of crop varieties, including bread wheat, from their wild ancestors. Explain the role of special genetic events, such as polyploidy, in the development of many modern crop varieties. 275 276







Applications of gene technologies

Learning aims and skills

Activity number

☐ 3 Explain how humans use gene technologies to alter selection pressures and thereby intervene in the evolutionary process. Examples include:

(a) The application of gene cloning in gene research and in the production of valuable commodities (see b). 143

(b) Use of genetically engineered bacteria (bacterial transformation) to express large quantities of a gene product (e.g. antibiotics, human insulin). 157 158

(c) The use of DNA profiling and genetic screening in the diagnosis and future management of heritable disorders. 277

(d) Use of gene therapy to treat certain diseases (e.g. SCIDS, cystic fibrosis). Understand and explain the role of vectors in gene therapy treatment. 278 279

(e) Use of stem cell technology (including therapeutic cloning) to produce cell lines for research and specific immunocompatible tissues to treat human diseases, e.g. Stargardt's disease and diabetes. 280 281

The future of human evolution

Learning aims and skills

Activity number

☐ 4 Describe how human intervention (e.g. advances in medical technology) can alter the selection pressures currently acting on the human population. 282



273 Selective Breeding in Animals

Key Idea: Selective breeding of domesticated animals involves identifying desirable qualities (e.g. high wool production or meat yield) and breeding together individuals with those qualities so the trait is reliably passed on.

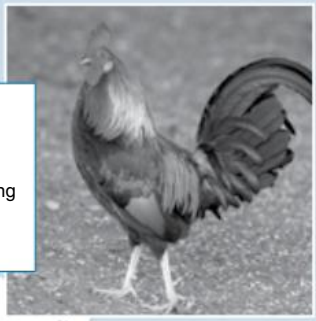
The domestication of livestock dates back at least 8000 years. Today's important stock breeds were all derived from wild ancestors that were domesticated by humans, who then used **selective breeding** to produce livestock to meet specific requirements. Practices such as **inbreeding**, **line-breeding**, and **out-crossing** are used to select and 'fix' desirable traits in varieties. Today, modern breeding techniques often employ reproductive technologies, such as artificial insemination, so that the desirable characteristics of one male can be passed on to many offspring. These new technologies refine the selection process and increase the rate at which stock improvements are made. Rates are predicted to accelerate further as new technologies, such as genomic selection, become more widely available and less costly. However, producing highly inbred lines of animals with specific traits can have disadvantages. **Homozygosity** for a number of desirable traits can create welfare issues, e.g. excessively rapid weight gain can cause joint deformities.

The Origin of Domestic Animals



PIG
Wild ancestor: Boar
Origin: Anatolia, 9000 years BP
Now: More than 12 distinct modern breeds, including the Berkshire (meat) and Tamworth (hardiness).

DOMESTIC FOWL
Wild ancestor: Red jungle fowl
Origin: Indus Valley, 4000 BP
Now: More than 60 breeds including Rhode Island Red (meat) and Leghorn (egg production).



Each domesticated breed has been bred from the wild ancestor (pictured). The date indicates the earliest record of the domesticated form (years before present or BP). Different countries have different criteria for selection, based on their local environments and consumer preferences.







GOAT
Wild ancestor: Bezoar goat
Origin: Iraq, 10,000 years BP
Now: approx. 35 breeds including Spanish (meat), Angora (fibre) and Nubian (dairy).

SHEEP
Wild ancestor: Asiatic mouflon
Orign: Iran, Iraq, Levant, 10,000 years BP
Now: More than 200 breeds including Merino (wool), Suffolk (meat), Friesian (milk), and dual purpose (Romney).

CATTLE
Wild ancestor: Auroch (extinct)
Origin: SW Asia, 10,000 years BP
Now: 800 modern breeds including the Aberdeen angus (meat), Friesian and Jersey (milk), and Zebu (draught).

1. Distinguish between inbreeding and out-crossing, explaining the significance of each technique in selective breeding:

2. How are new reproductive technologies contributing to rapid phenotypic change in populations?

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Dogs provide a striking example of selective breeding, with more than 400 recognised breeds. Over centuries, humans have selected for desirable physical and behavioural traits. All breeds of dog are members of the same species, ***Canis familiaris***. This species descended from a single wild species, the grey wolf ***Canis lupus***, over 15,000 years ago. Five ancient dog breeds are recognised, from which all other breeds are thought to have descended by selective breeding.

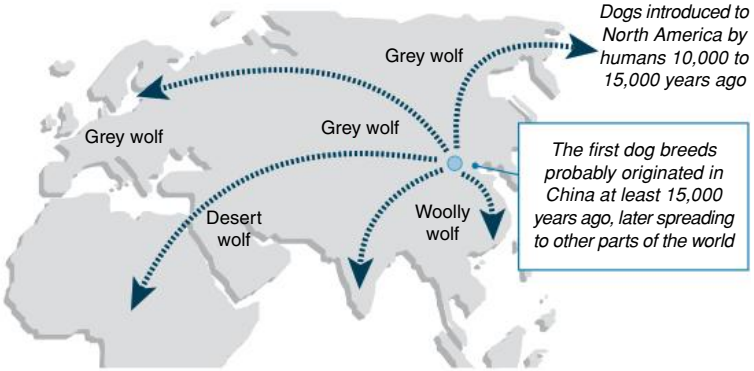


Grey wolf *Canis lupus pallipes*

The grey wolf is distributed throughout Europe, North America, and Asia. Amongst members of this species, there is a lot of variation in coat coloration. This accounts for the large variation in coat colours of dogs today.

The Ancestor of Domestic Dogs

Until recently, it was unclear whether the ancestor to the modern domestic dogs was the desert wolf of the Middle East, the woolly wolf of central Asia, or the grey wolf of Northern Hemisphere. Recent genetic studies (mitochondrial DNA comparisons) now provide strong evidence that the ancestor of domestic dogs throughout the world is the grey wolf. It seems likely that this evolutionary change took place in a single region, most probably China.





Mastiff-type
Canis familiaris inostranzevi
Originally from Tibet, the first records of this breed of dog go back to the Stoneage.



Greyhound
Canis familiaris leineri
Drawings of this breed on pottery dated from 8000 years ago in the Middle East make it one of the oldest.



Pointer-type
Canis familiaris intermedius
Probably derived from the greyhound breed for the purpose of hunting small game.



Sheepdog
Canis familiaris metris optimae
Originating in Europe, this breed has been used to guard flocks from predators for thousands of years.



Wolf-like
Canis familiaris palustris
Found in snow covered habitats in northern Europe, Asia (Siberia), and North America (Alaska).

3. How has selective breeding contributed to changes in the gene pool of domestic dogs? _____

4. Which behavioural characteristics of wolves predisposed them to successful domestication? _____

5. List the physical and behavioural traits that would be desirable (selected for) in the following uses of a dog:

   (a) Hunting large game (e.g. boar and deer): _____

   (b) Game fowl dog: _____

   (c) Stock control (sheep/cattle dog): _____

   (d) Family pet (house dog): _____

   (e) Guard dog: _____

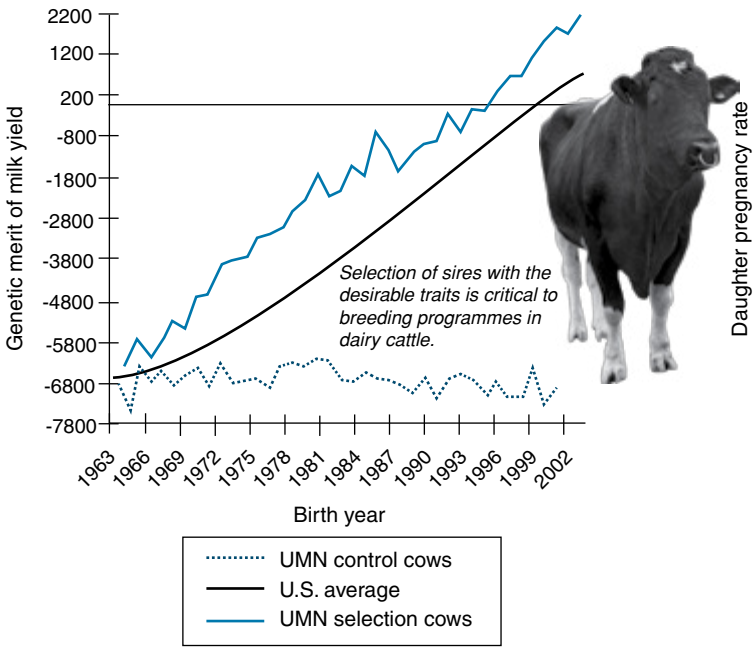
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274 Selection and Population Change

Key Idea: Selective breeding is a method for rapidly producing change in the phenotypic characteristics of a population.

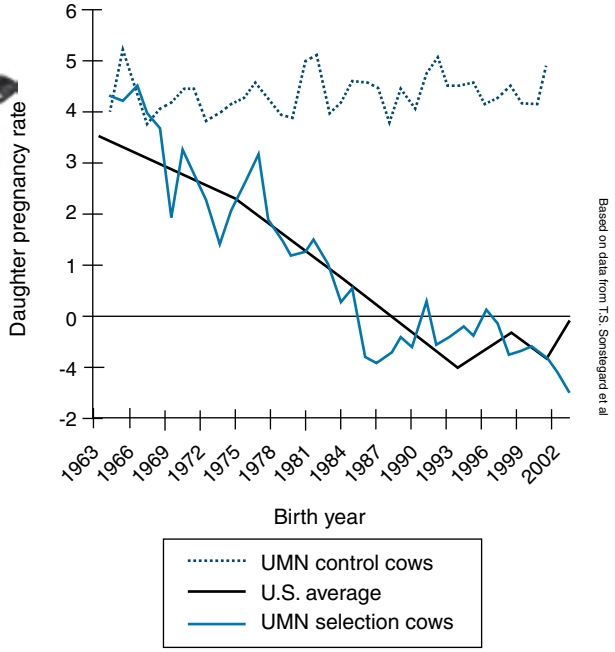
Humans may create the selection pressure for evolutionary change by choosing and breeding together individuals with particular traits. The example of milk yield in Holstein cows (below) illustrates how humans have directly influenced the genetic makeup of Holstein cattle with respect to milk production and fertility. Since the 1960s, the University of Minnesota has maintained a Holstein cattle herd that has not been subjected to any selection. They also maintain a herd that was subjected to selective breeding for increased milk production between 1965 and 1985. They compared the genetic merit of milk yield in these groups to that of the USA Holstein average.

Gain in Genetic Merit of Milk Yield



Milk production in the University of Minnesota herd subjected to selective breeding increased in line with the U.S. average production. In real terms, milk production per cow per milking season increased by 3740 kg since 1964. The herd with no selection remained effectively constant for milk production.

Fertility in Holstein Cows



Along with increased milk production there has been a distinct decrease in fertility. The fertility of the University of Minnesota herd that was not subjected to selection remained constant while the fertility of the herd selected for milk production decreased with the U.S. fertility average.

1. (a) Describe the relationship between milk yield and fertility on Holstein cows: _____

   _____

   (b) What does this suggest about where the genes for milk production and fertility are carried? _____

   _____

   _____

2. What limits might this place on maximum milk yield? _____

   _____

3. Why is sire selection important in selective breeding, even if the characters involved are expressed only in the female?

   _____

4. Natural selection is the mechanism by which organisms with favourable traits become proportionally more common in the population. How does selective breeding mimic natural selection? How does the example of the Holstein cattle show that reproductive success is a compromise between many competing traits?

   _____

   _____

   _____

   _____

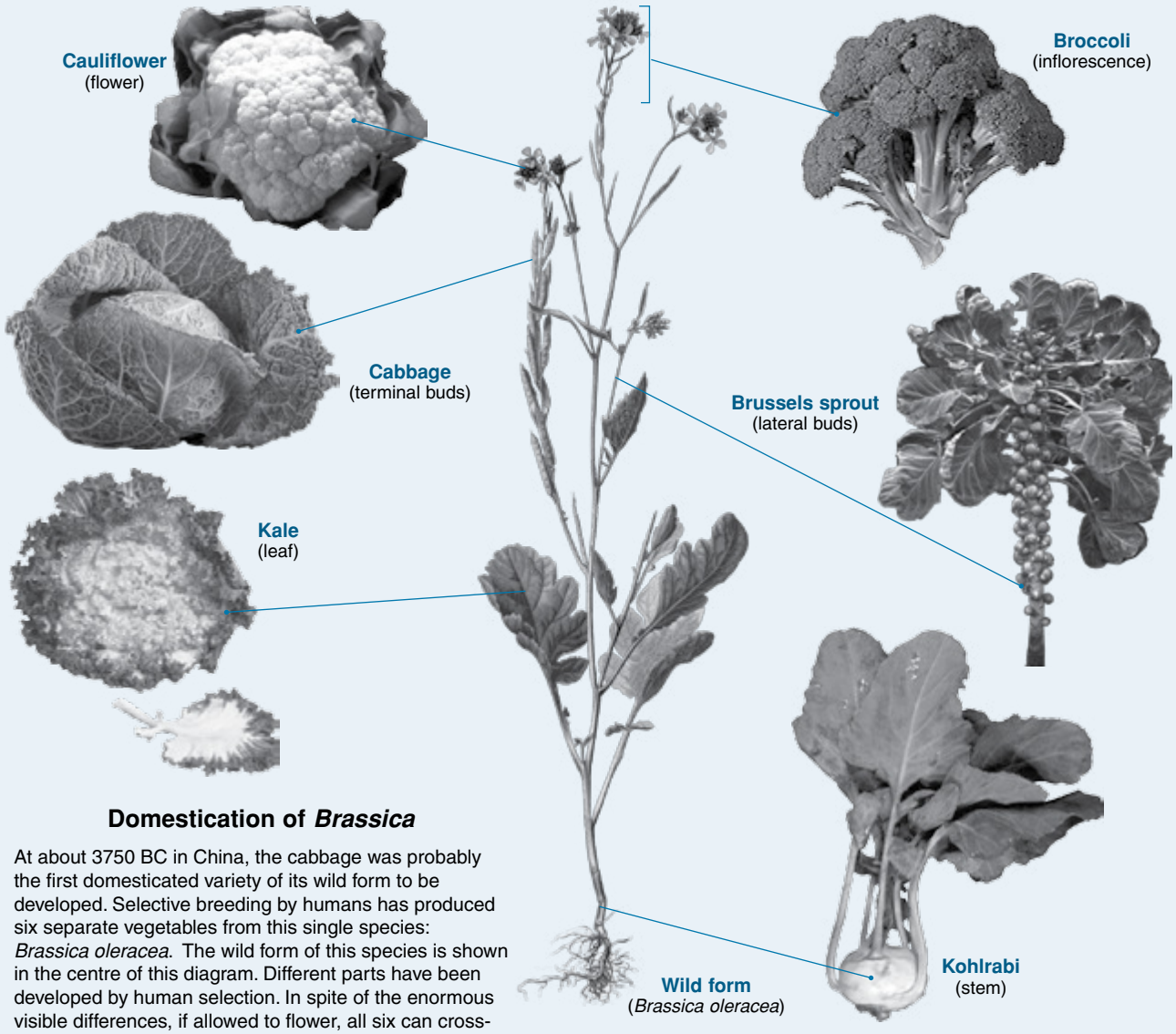
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275 Selective Breeding in Crop Plants

Key Idea: The genetic diversity within crop varieties provides options to develop, through selection, new crop plants.

For thousands of years, farmers have used the variation in wild and cultivated plants to develop crops. *Brassica oleracea* is a good example of the variety that can be produced by selectively growing plants with desirable traits. Not only are there six varieties of *Brassica oleracea*, but each of those has a number of sub-varieties as well. Although brassicas have been cultivated for several thousand years, cauliflower, broccoli, and brussels sprouts appeared only in the last 500 years.



Domestication of *Brassica*

At about 3750 BC in China, the cabbage was probably the first domesticated variety of its wild form to be developed. Selective breeding by humans has produced six separate vegetables from this single species: *Brassica oleracea*. The wild form of this species is shown in the centre of this diagram. Different parts have been developed by human selection. In spite of the enormous visible differences, if allowed to flower, all six can cross-pollinate. Kale is closer to the wild type than the other related breeds.

1. Study the diagram above and identify which part of the plant has been selected for to produce each of the vegetables:

   (a) Cauliflower: _____ (d) Brussels sprout: _____

   (b) Kale: _____ (e) Cabbage: _____

   (c) Broccoli: _____ (f) Kohlrabi: _____

2. Describe the feature of these vegetables that suggests they are members of the same species: _____

   _____

3. Describe the method used to develop broccoli and the features one would look for when doing so: _____

   _____

   _____

   _____

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USDA

The number of apple varieties is now a fraction of the many hundreds grown a century ago. Apples are native to Kazakhstan and breeders are now looking back to this centre of diversity to develop apples resistant to the bacterial disease that causes fire blight.



In 18th-century Ireland, potatoes were the main source of food for about 30% of the population, and farmers relied almost entirely on one very fertile and productive variety. That variety proved susceptible to the potato blight fungus which resulted in a widespread famine.



Hybrid corn varieties have been bred to minimise damage by insect pests such as corn rootworm (above). Hybrids are important because they recombine the genetic characteristics of parental lines and show increased heterozygosity and hybrid vigour.

4. The genetic processes involved in selective breeding and natural selection are essentially no different. Explain how this has changed with the advent of genetic engineering technology and why it is particularly relevant to crop plants:

5. Describe a phenotypic characteristic that might be desirable in an apple tree and explain your choice:

6. (a) Explain why genetic diversity might decline during selective breeding for particular characteristics:

   (b) With reference to an example, discuss why retaining genetic diversity in crop plants is important for food security:

7. Cultivated American cotton plants have a total of 52 chromosomes (2N = 52). In each cell there are 26 large chromosomes and 26 small chromosomes. Old World cotton plants have 26 chromosomes (2N = 26), all large. Wild American cotton plants have 26 chromosomes, all small. How might cultivated American cotton have originated from Old World cotton and wild American cotton:

8. The Cavendish is the variety of banana most commonly sold in world supermarkets. It is seedless, sterile, and under threat of extinction by Panama disease Race 4. Explain why Cavendish banana crops are so endangered by this fungus:

9. Discuss the need to maintain the biodiversity of wild plants and ancient farm breeds:

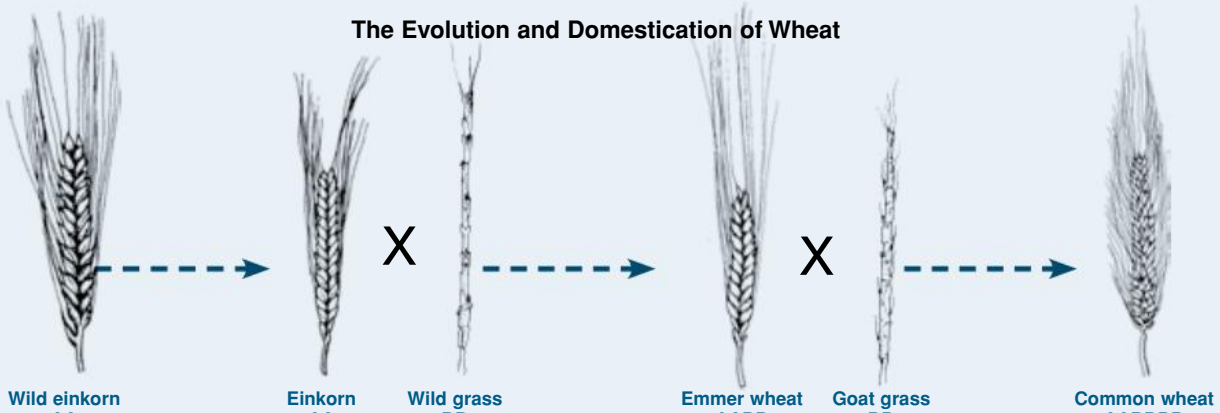
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276 Breeding Modern Wheat

Key Idea: Modern wheat evolved as a result of two natural hybridisation events and the doubling of its chromosomes.

Wheat has been cultivated for more than 9000 years and has undergone many genetic changes during its domestication. The evolution of modern bread wheat from its wild ancestors (below) involved two natural **hybridisation** events, accompanied by **polyploidy**. Once wheat became domesticated, selective breeding emphasised characteristics such as high protein (gluten) content, high yield, and pest and disease resistance. Hybrid vigour in wheat cultivars is produced by crossing inbred lines and selecting for desirable traits in the progeny, which can now be identified using genetic techniques such as marker assisted selection. This is an indirect selection process where a trait of interest is selected on the basis of a marker linked to it. Increasingly, research is focused on enhancing the genetic diversity of wheat to provide for future crop development. With this in mind, there is renewed interest in some of the lower yielding, ancient wheat varieties, which possess alleles no longer present in modern inbred varieties.

The Evolution and Domestication of Wheat



Wild einkorn becomes domesticated in the Middle East. There are slight changes to phenotype but not chromosome number.

A sterile hybrid between einkhorn and wild grass undergoes a chromosome doubling to create fertile emmer wheat.

A sterile hybrid between emmer wheat and goat grass undergoes a chromosome doubling to create fertile common wheat.



Ancient cereal grasses had heads which shattered easily so that the seeds were widely scattered. In this more primitive morphology, the wheat ear breaks into spikelets when threshed, and milling or pounding is needed to remove the hulls and obtain the grain. Cultivation and repeated harvesting and sowing of the grains of wild grasses led to domestic strains with larger seeds and sturdier heads. Modern selection methods incorporate genetic techniques to identify and isolate beneficial genes, e.g. the RHt dwarfing gene, which gave rise to shorter stemmed modern wheat varieties.



Modern bread wheat has been selected for its non-shattering heads, high yield, and high gluten (protein) content. The grains are larger and the seeds (spikelets) remain attached to the ear by a toughened rachis during harvesting. On threshing, the chaff breaks up, releasing the grains. Selection for these traits by farmers might not necessarily have been deliberate, but occurred because these traits made it easier to gather the seeds. Such '**incidental' selection** was an important part of crop domestication. **Hybrid vigour** in cultivars is generated by crossing inbred lines.



Durum wheat is a modern variety developed by selective breeding of the domesticated emmer wheat strains. Durum (also called hard wheat) has large firm kernels with a high protein content. These properties make it suitable for pasta production. As with all new wheat varieties, new cultivars are produced by crossing two lines using hand pollination, then selfing or **inbreeding** the progeny that combine the desirable traits of both parents. Progeny are evaluated for several years for the traits of interest, until they can be released as established varieties or cultivars.

1. Describe three phenotypic characteristics that would be desirable in a wheat plant:

   (a) _____

   (b) _____

   (c) _____

2. Explain how both natural events and selective breeding have contributed to the high yielding modern wheat varieties:

   _____

   _____

KNOW WEB 276 LINK 237 LINK 275

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277 A Gene That Can Tell Your Future



Key Idea: Huntington's disease is caused by a repeating section of DNA. The longer the repeating pattern, the earlier the disease tends to appear and the worse its symptoms.

Huntington's disease (HD) is a genetic neuro-degenerative disease that normally does not affect people until about the age of 40. Its symptoms usually appear first as a shaking of the hands and an awkward gait. Later manifestations of the disease include serious loss of muscle control and mental function, often ending in dementia and ultimately death.

All humans have the huntingtin (**HTT**) gene, which in its normal state produces a protein with roles in gene transcription, synaptic transmission, and brain cell survival. The mutant gene (**mHTT**) causes changes to and death of the cells of the cerebrum, the hippocampus, and cerebellum, resulting in the atrophy (reduction) of brain matter. The gene was discovered by Nancy Wexler in 1983 after ten years of research working with cell samples and family histories of more than 10,000 people from the town of San Luis in Venezuela, where around 1% of the population have the disease (compared to about 0.01% in the rest of the world). Ten years later the exact location of the gene on the chromosome 4 was discovered.

The identification of the HD gene began by looking for a gene probe that would bind to the DNA of people who had HD, and not to those who didn't. Eventually a marker for HD, called **G8**, was found. The next step was to find which chromosome carried the marker and where on the chromosome it was. The researchers hybridised human cells with those of mice so that each cell contained only one human chromosome, a different chromosome in each cell. The hybrid cell with chromosome 4 was the one with the G8 marker. They then found a marker that overlapped G8 and then another marker that overlapped that marker. By repeating this many times, they produced a map of the genes on chromosome 4. The researchers then sequenced the genes and found people who had HD had one gene that was considerably longer than people who did not have HD, and the increase in length was caused by the repetition of the base sequence CAG.

The HD mutation (mHTT) is called a trinucleotide repeat expansion. In the case of mHTT, the base sequence CAG is repeated multiple times on the short arm of chromosome 4. The normal number of CAG repeats is between 6 and 30. The mHTT gene causes the repeat number to be 35 or more and the size of the repeat often increases from generation to generation, with the severity of the disease increasing with the number of repeats. Individuals who have 27 to 35 CAG repeats in the HTT gene do not develop Huntington disease, but they are at risk of having children who will develop the disorder. The mutant allele, mHTT, is also dominant, so those who are homozygous or heterozygous for the allele are both at risk of developing HD.

New research has shown that the mHTT gene activates an enzyme called JNK3, which is expressed only in the neurones and causes a drop in nerve cell activity. While a person is young and still growing, the neurones can compensate for the accumulation of JNK3. However, when people get older and neurone growth stops, the effects of JNK3 become greater and the physical signs of HD become apparent. Because of mHTT's dominance, an affected person has a 50% chance of having offspring who are also affected. Genetic testing for the disease is relatively easy now that the genetic cause of the disease is known. While locating and counting the CAG repeats does not give a date for the occurrence of HD, it does provide some understanding of the chances of passing on the disease.



NYWTS

American singer-songwriter and folk musician Woody Guthrie died from complications of HD

1. Describe the physical effects of Huntington's disease: _____

2. Describe how the mHTT gene was discovered: _____

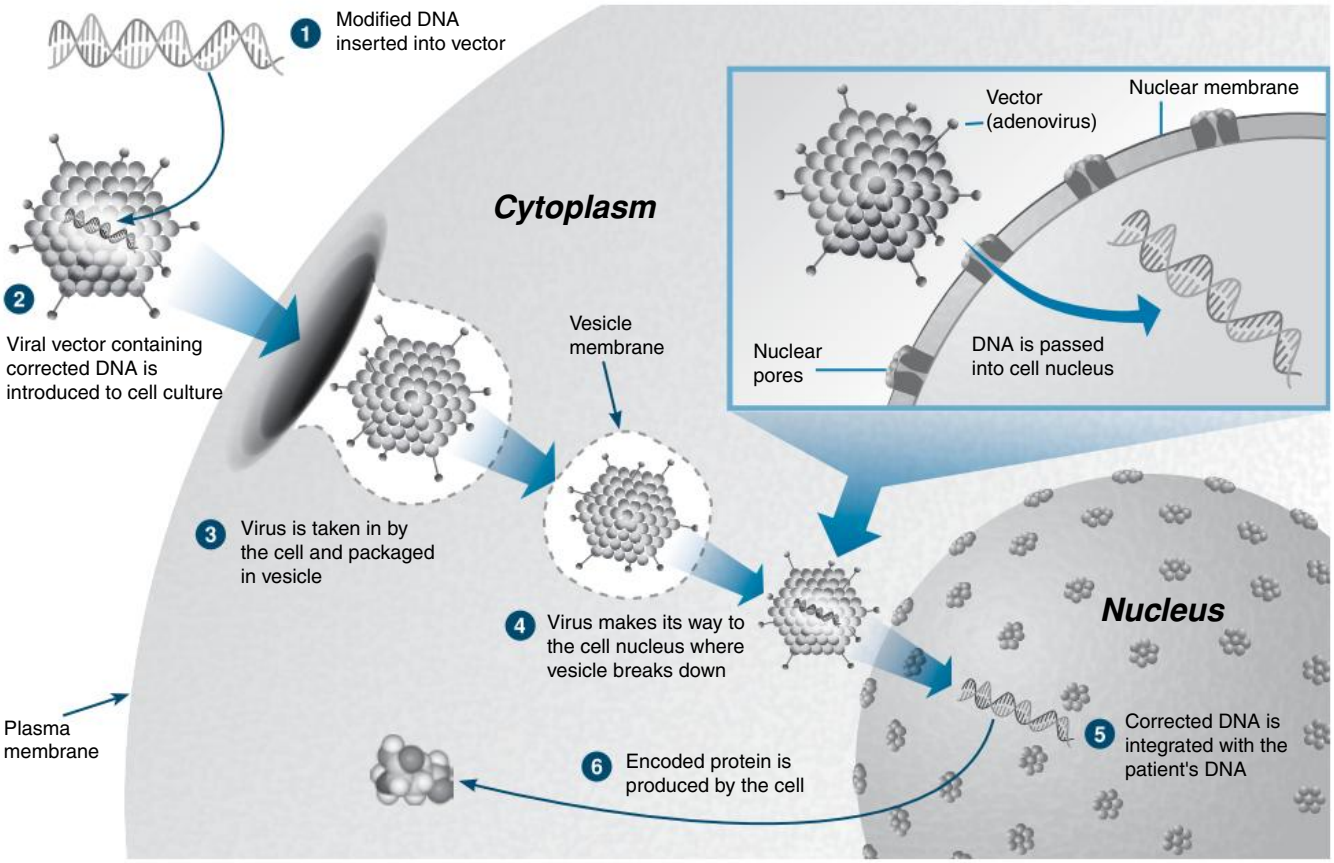
3. Discuss the cause of Huntington's disease and its pattern of increasing severity with each generation: _____

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278 Gene Therapy

Key Idea: Gene therapy aims to replace faulty genes by using a vector to transfer the correctly functioning gene into a patient's DNA.

Gene therapy uses gene technology to treat disease by correcting or replacing faulty genes. Although the details vary, all gene therapies are based around the same technique. The correct non-faulty gene is inserted into a vector, a carrier which transfers the DNA into the patient's cells (**transfection**). The vector is introduced into a sample of the patient's cells, and these are cultured to amplify the correct gene. The cultured cells are then transferred back to the patient. The use of altered stem cells instead of mature somatic cells has achieved longer lasting results in many patients. The treatment of somatic cells or stem cells is therapeutic (provides a benefit) but the changes are not inherited. **Germline therapy** (modification of the gametes) would enable genetic changes to be passed on. Gene therapy has had limited success because transfection of targeted cells is inefficient, and the side effects can be severe or even fatal. However, gene therapy to treat SCID, a genetic disease affecting the immune system, has had some success.



1. (a) Describe the general principle of gene therapy: _____

_____

_____

(b) Describe the medical areas where gene therapy might be used: _____

_____

_____

2. Explain the significance of transfecting **germline cells** rather than **somatic cells**: _____

_____

_____

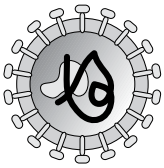
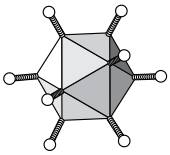
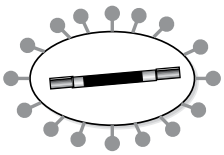
_____

3. Explain the purpose of **gene amplification** in gene therapy: _____

_____

_____

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	Vectors That Can Be Used For Gene Therapy			
	 **Retrovirus**	 **Adenovirus**	 **Liposome**	 **Naked DNA**
Insert size:	8000 bases	8000 bases	>20,000 bases	>20,000 bases
Integration:	Yes	No	No	No
***In vivo* delivery:**	Poor	High	Variable	Poor
Advantages	• Integrate genes into the chromosomes of the human host cell. • Offers chance for long-term stability.	• Modified for gene therapy, they infect human cells and express the normal gene. • Most do not cause disease. • Have a large capacity to carry foreign genes.	• Liposomes seek out target cells using sugars in their membranes that are recognised by cell receptors. • Have no viral genes that may cause disease.	• Have no viral genes that may cause disease. • Expected to be useful for vaccination.
Disadvantages	• Many infect only cells that are dividing. • Genes integrate randomly into chromosomes, so might disrupt useful genes in the host cell.	• Viruses may have poor survival due to attack by the host's immune system. • Genes may function only sporadically because they are not integrated into host cell's chromosome.	• Less efficient than viruses at transferring genes into cells, but recent work on using sugars to aid targeting have improved success rate.	• Unstable in most tissues of the body. • Inefficient at gene transfer.

In the table above, the following terms are defined as follows: **Naked DNA**: the genes are applied by ballistic injection (firing using a gene gun) or by regular hypodermic injection of plasmid DNA. **Insert size**: size of gene that can be inserted into the vector. **Integration**: whether or not the gene is integrated into the host DNA (chromosomes). ***In vivo* delivery**: ability to transfer a gene directly into a patient.

4. Explain why genetically modified stem cells offer greater potential in gene therapy treatments than GM somatic cells:

5. (a) Describe the features of viruses that make them well suited as **vectors** for gene therapy:

   (b) Describe two problems with using viral vectors for gene therapy:

6. (a) Explain why it may be beneficial for a (therapeutic) gene to integrate into the patient's chromosome:

   (b) Explain why this has the potential to cause problems for the patient:

7. (a) Explain why naked DNA is likely to be unstable within a patient's tissues:

   (b) Explain why enclosing the DNA within liposomes might provide greater stability:

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279 Gene Delivery Systems

Key Idea: The delivery of genes into target cells and then into patients has proved technically difficult, limiting the use of gene therapy.

It remains technically difficult to deliver genes successfully to a patient, limiting the success rate of gene therapy treatments. Any improvements have been mostly short-lived, or counteracted by adverse side effects. The inserted genes may reach only about 1% of target cells. Those that reach their target may work inefficiently and produce too little protein, too slowly to be of benefit. Many patients also have immune reactions to the vectors used in gene transfer. One of the first gene therapy trials was for cystic fibrosis (CF). CF was an obvious candidate for gene therapy because, in most cases, the disease is caused by a single, known gene mutation. However, despite its early promise, gene therapy for this disease has been disappointing (below right). Severe Combined Immune Deficiency (SCID) is another candidate for gene therapy, again because the disease is caused by single, known mutation (below left). Gene therapies for this disease have so far proved promising.

Treating SCID using Gene Therapy

The most common form of **SCID** (Severe Combined Immune Deficiency) is **X-linked SCID**, which results from mutations to a gene on the X chromosome encoding the **common gamma chain**, a protein forming part of a receptor complex for numerous types of leucocytes. A less common form of the disease, (**ADA-SCID**) is caused by a defective gene that codes for the enzyme adenosine deaminase (ADA).

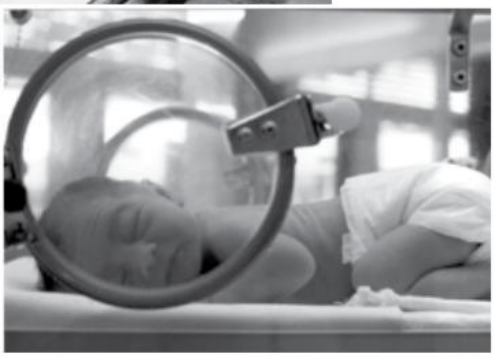
Both of these types of SCID lead to immune system failure. A common treatment for SCID is bone marrow transplant, but this is not always successful and runs the risks of infection from unscreened viruses. **Gene therapy** appears to hold the best chances of producing a cure for SCID because the mutation affects only one gene whose location is known. DNA containing the corrected gene is placed into a **gutted retrovirus** and introduced to a sample of the patient's **bone marrow.** The treated cells are then returned to the patient.

In some patients with ADA-SCID, treatment was so successful that supplementation with purified ADA was no longer required. The treatment carries risks though. In early trials, two of ten treated patients developed leucaemia when the corrected gene was inserted next to a gene regulating cell growth.



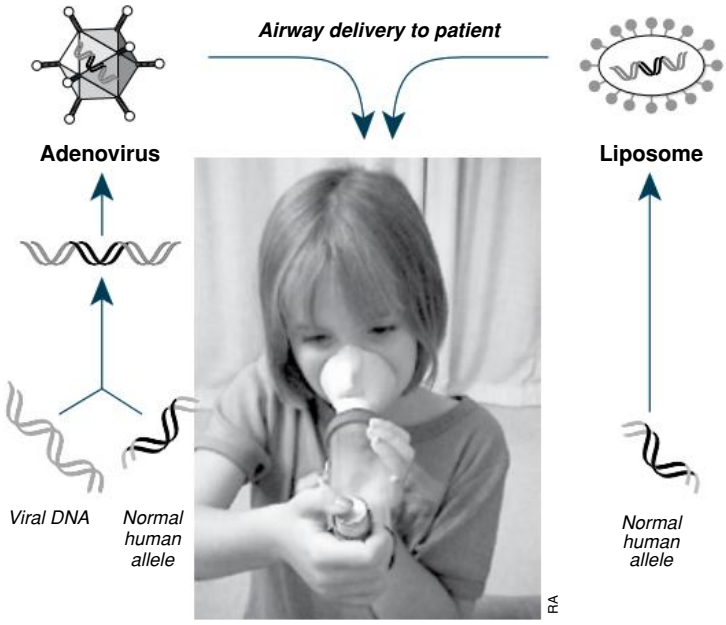
Georgetown University Hospital

Samples of bone marrow being extracted prior to treatment with gene therapy.



Jacoplane

Detection of SCID is difficult for the first months of an infant's life due to the mother's antibodies being present in the blood. Suspected SCID patients must be kept in sterile conditions at all times to avoid infection.



An **adenovirus** that normally causes colds is genetically modified to make it safe and to carry the normal (unmutated) CFTR ('cystic fibrosis') gene.

Liposomes are tiny fat globules. Normal CF genes are enclosed in liposomes, which fuse with plasma membranes and deliver the genes into the cells.

Gene Therapy - Potential Treatment for Cystic Fibrosis?

Cystic fibrosis (CF) is caused by a mutation to the gene coding for a chloride ion channel important in creating sweat, digestive juices, and mucus. The dysfunction results in abnormally thick, sticky mucus that accumulates in the lungs and intestines. The identification and isolation of the CF gene in 1989 meant that scientists could look for ways in which to correct the genetic defect rather than just treating the symptoms using traditional therapies.

The main target of CF gene therapy is the lung, because the progressive lung damage associated with the disease is eventually lethal.

In trials, normal genes were isolated and inserted into patients using vectors such as **adenoviruses** and **liposomes**, delivered via the airways (left). The results of trials were disappointing: on average, there was only a 25% correction, the effects were short lived, and the benefits were quickly reversed. Alarmingly, the adenovirus used in one of the trials led to the death of one patient.

Source: Cystic Fibrosis Trust, UK.

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1. A great deal of current research is being devoted to discovering a gene therapy solution to treat **cystic fibrosis** (CF):

   (a) Describe the symptoms of CF: _____

   (b) Why has this particular genetic disease been so eagerly targeted by geneticists? _____

   (c) Outline some of the problems so far encountered with gene therapy for CF: _____

2. Identify two vectors for introducing healthy CFTR genes into CF patients.

   (a) Vector 1: _____

   (b) Vector 2: _____

3. (a) Describe the difference between X-linked SCID and ADA-SCID: _____

   (b) Identify the vector used in the treatment of SCID: _____

4. Briefly outline the differences in the gene therapy treatment of CF and SCID: _____

5. Changes made to chromosomes as a result of gene therapy involving somatic cells are not inherited. Germ-line gene therapy has the potential to cure disease, but the risks and benefits are still not clear. For each of the points outlined below, evaluate the risk of germ-line gene therapy relative to somatic cell gene therapy and explain your answer:

   (a) Chance of interfering with an essential gene function: _____

   (b) Misuse of the therapy to selectively alter phenotype: _____

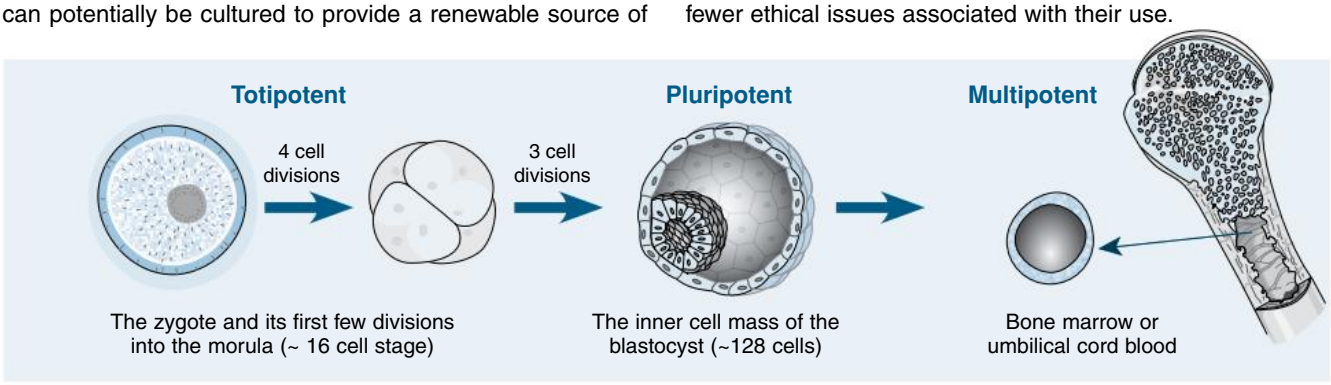


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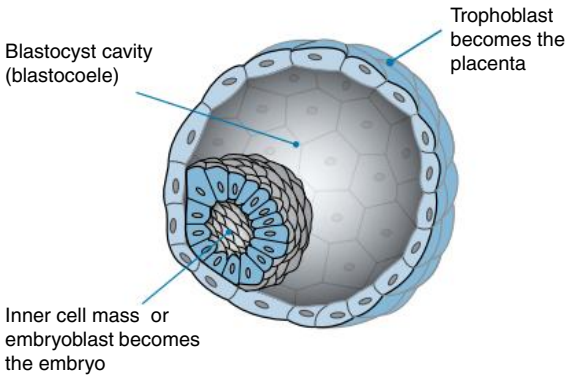
280 Types of Stem Cells

Key Idea: The potency of stem cells depends on their origin. Both embryonic and adult stem cells can be used to replace diseased and damaged tissue.

The properties of self renewal and potency make stem cells suitable for a wide range of applications. Stem cells from early stage embryos (embryonic stem cells) are pluripotent and can potentially be cultured to provide a renewable source of cells for studies of human development and gene regulation, for tests of new drugs and vaccines, for monoclonal antibody production, and for treating any type of diseased or damaged tissue. Adult stem cells from bone marrow or umbilical cord blood can give rise to a more limited number of cell types. Although their potential use is more restricted, there are fewer ethical issues associated with their use.

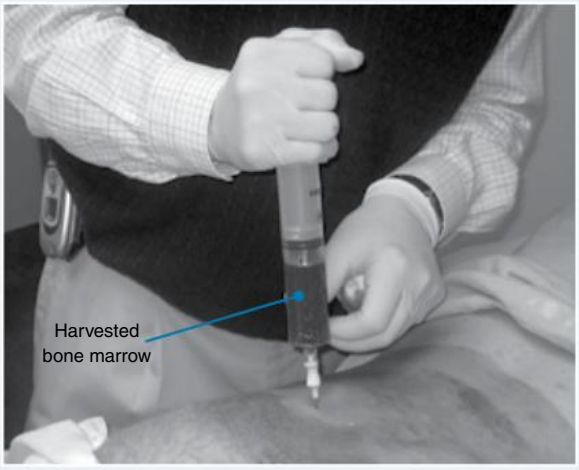


Embryonic Stem Cells



Embryonic stem cells (**ESC**) are derived from the inner cell mass of blastocysts (above). Blastocysts are embryos that are about five days old and consist of a hollow ball of 50-150 cells. Cells derived from the inner cell mass are **pluripotent**. They can become any cells of the body, with the exception of placental cells. When cultured without any stimulation to differentiate, ESC retain their potency through multiple cell divisions. This means they have great potential for therapeutic use in regenerative medicine and tissue replacement. However, the use of ESC involves the deliberate creation and destruction of embryos and is therefore is ethically unacceptable to many people.

Adult Stem Cells



Andrew Raito Stevenfruitsmaak

Adult stem cells (ASC) are undifferentiated cells found in several types of tissues (e.g. brain, bone marrow, fat, and liver) in adults, children, and umbilical cord blood. Unlike ESCs, they are **multipotent** and can only differentiate into a limited number of cell types, usually related to the tissue of origin. There are fewer ethical issues associated with using ASC for therapeutic purposes, because no embryos are destroyed. For this reason, ASC are already widely used to treat a number of diseases including leukemia and other blood disorders.

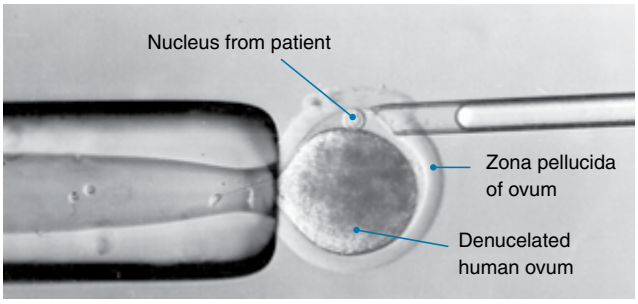
1. (a) Distinguish between embryonic stem cells and adult stem cells with respect to their **potency**:

(b) What is the significance of this difference to their use in the treatment of disease:

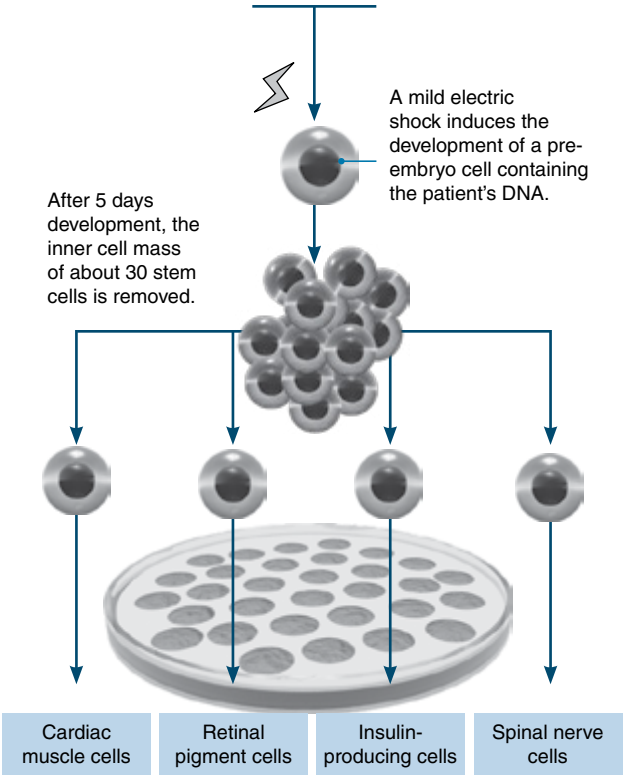
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Embryonic Stem Cell (ESC) Cloning

ESC can come from embryos that have been fertilized *in vitro* and then donated for research. These cell lines will not be patient-matched because each new embryo is unique. However, ESC can also come from cloned embryos created using somatic cell nuclear transfer using a donor nucleus from the patient, as shown below. These ESC lines will patient-matched.



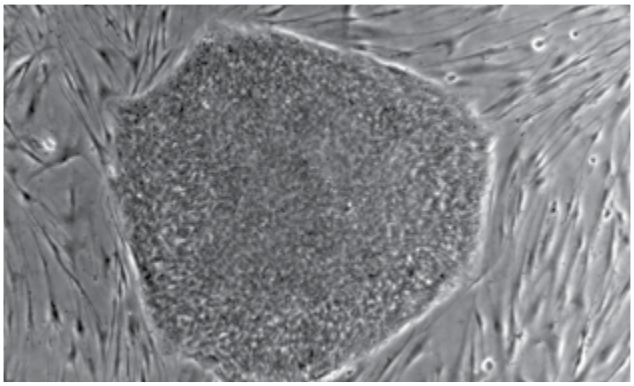
DW



When ESCs are provided with appropriate growth factors and conditions, they will differentiate into specific specialized cell types.

Issues in ESC Cloning

For all tissue transplants, e.g. blood transfusions and bone marrow transplants, tissues must be matched for histocompatibility between different individuals. If donor material is poorly matched to the recipient, the recipient's immune system rejects the donor cells. Stem cell cloning (**therapeutic cloning**) provides a way around this problem. Stem cell cloning produces genetically matched stem cells that can be turned into any cell type in the human body.



NIH

Human embryonic stem cells (hESC) growing on mouse embryonic fibroblasts. The mouse fibroblasts act as feeder cells for the culture, releasing nutrients and providing a surface for the ESCs to grow on.

ESC therapy has enormous potential to make life changing improvements to the health of people with diseased or damaged organs. Organs or tissues derived from a patient's ESC could be transplanted back into that patient without fear of tissue rejection or the need for ongoing immunosuppressive drug therapies. Despite this, many groups oppose the use of therapeutic cloning for many reasons including:

- The technology used to create the embryo could be used for reproductive cloning, i.e. creating a clone of the original human.
- The creation of stem cell line requires the destruction of a human embryo and thus human life.
- Human embryos have the potential to develop into an individual and thus have the same rights of the individual.
- Saving or enhancing the quality of life of an individual does not justify the destruction of the life of another (i.e. the embryo).
- ESC research has not produced any viable long term treatment, while other techniques (e.g. adult stem cells) have.
- There are other stem cell techniques that do not require the creation of an embryo but achieve similar results (e.g. cell lines grown from adult stem cells or umbilical cord blood).

2. (a) In your opinion, what is the one most important ethical issue associated with the use of ESC in medicine?

(b) What advantage does therapeutic cloning offer over conventional therapeutic use of embryonic stem cells?

3. Umbilical cord blood is promoted as a rich source of multipotent stem cells for autologous (self) transplants. Can you see a problem with the use of a baby's cord blood to treat a disease in that child at a later date?

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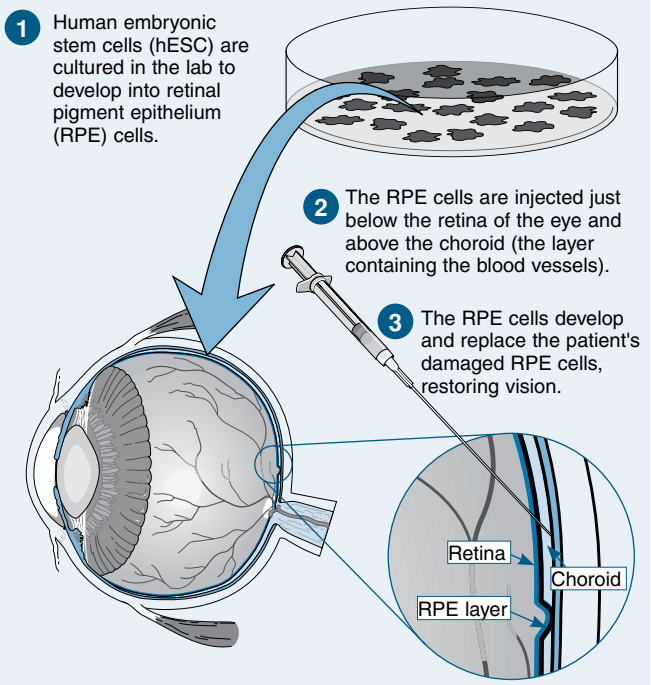
281 Using Stem Cells to Treat Disease

Key Idea: Embryonic stem cells have been used to treat Stargardt's disease with apparent success. New techniques make it possible to produce pluripotent cells for widespread therapeutic use in an ethically acceptable way.

The therapeutic use of embryonic stem cells (ESC) to replace lost retinal cells in patients with Stargardt's disease (an eye disease) has shown that such therapies are able to restore function to diseased organs. Future treatments using induced pluripotent cells derived from adult tissues will create patient-matched cell lines and bypass the need for embryos.

Stem Cells for Stargardt's Disease

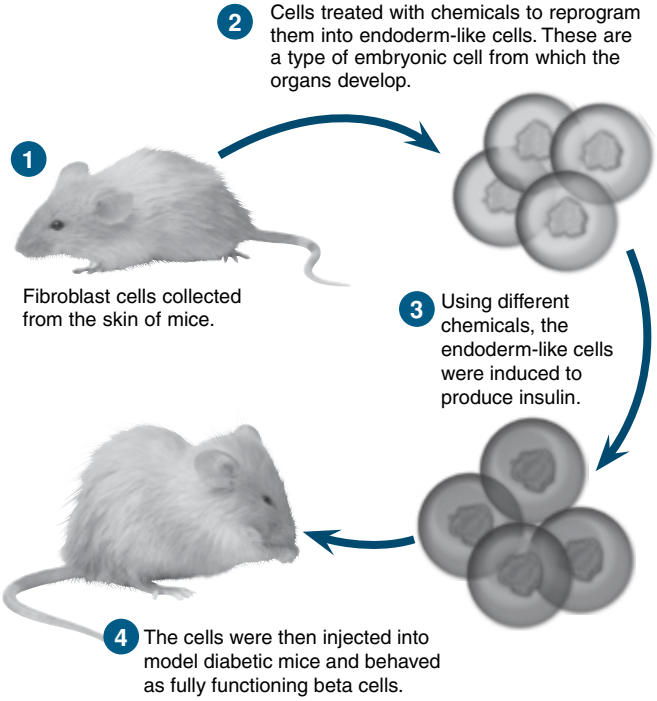
Stargardt's disease is an inherited form of juvenile macular degeneration (a loss of the central visual field of the eye). The disease is associated with a number of mutations and results in dysfunction of the retinal pigment epithelium (RPE) cells, which nourish the retinal photoreceptor cells and protect the retina from excess light. Dysfunction of the RPE causes deterioration of the photoreceptor cells in the central portion of the retina and progressive loss of central vision. This often begins between ages 6 and 12 and continues until a person is legally blind. Trials using stem cells have obtained promising results in treating the disease.



Stem Cells for Type 1 Diabetes?

Type 1 diabetes results from the body's own immune system attacking and destroying the insulin producing beta cells of the pancreas. In theory, new beta cells could be produced using stem cells. Research is focused on how to obtain the stem cells and deliver them effectively to the patient. Many different techniques are currently being investigated. Most techniques use stem cells from non-diabetics, requiring recipients to use immunosuppressant drugs so the cells are not rejected.

A study published in 2014 described a method for treating type 1 diabetes in mice using fibroblast cells taken from the skin of mice.



1. Describe one potential advantage of using embryonic stem cells for tissue engineering technology:

2. (a) Explain the basis for correcting Stargardt's disease using stem cell technology:

   (b) Suggest why researchers derived the RPE cells from embryos rather than by reprogramming a patient's own cells:

   (c) What advantage is there in reprogramming a patient's own cells and when would this be a preferable option?

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282 Present and Future Human Evolution

Key Idea: Human evolution is on-going, although the selections pressures experienced today are very different from those experienced by our ancestors.

The modern human environment is vastly different to that experienced by our human ancestors. It is common to link our modern lifestyles with increased rates of cancers and other diseases, but factors in our current environment can only act as selection pressures if they affect reproductive fitness. Some of the trends identified in the evolution of human anatomy (below) may continue, but new selection pressures may increasingly influence human evolution. In the last 200 years, rapid developments in science, technology, and medicine have helped humans to withstand and manipulate conditions that would otherwise have acted as selection pressures. Medical advancements and genetic technology have helped to lower mortality, increase fertility, and assist the survival of medically compromised offspring (e.g. premature babies and the offspring of multiple births). Even the ease of global travel adds a new selection pressure, as new diseases can be easily transferred across countries through air travel.

Evolutionary Trends

Brain: Increase in brain size and complexity.

Nasal form: A reduction in the width of the nose.

Face: Reduced facial projection leading to a flatter profile. There is a trend towards a smaller, more vertical face, with loss of brow ridges.

Dentition: Reduction in tooth size (particularly of the canines) and loss of the diastema.

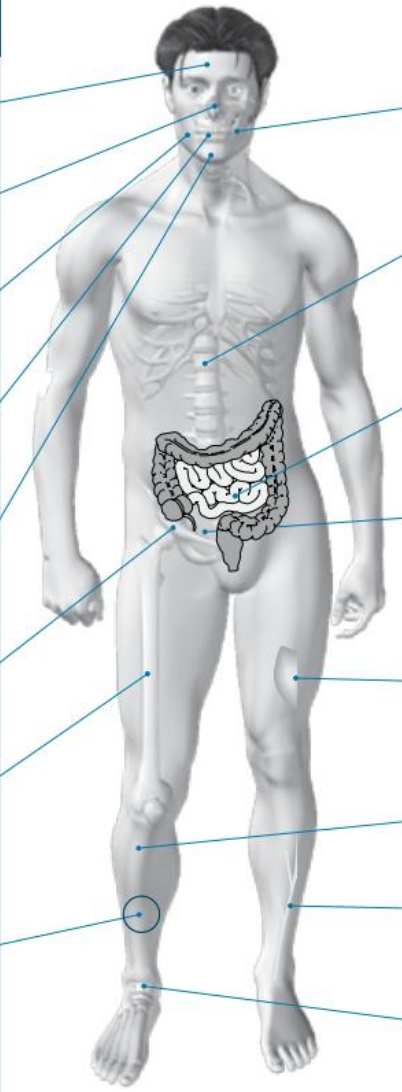
Jaw: Reduction in the size and robusticity of the jaw, with increasing loss of a prominent chin.

Pelvis: A trend towards a shorter and broader pelvis in order to accommodate the attachment of leg muscles adapted for walking.

Femur: Improvements in the adaptations of the legs (angle, stability, and strength of the femur) for upright bipedal locomotion.

Body hair: Reduction in the coarseness of body hair over much of the body surface (there has been no reduction in the number of hair follicles however).

After C.B. Stringer



Associated Problems

Teeth: Wisdom teeth become overcrowded and impacted as a result of reduced jaw size.

Slipped disc: Lower back troubles, usually the result of degenerative changes with age, are compounded by the load being carried by only two limbs.

Weakened muscles: Muscles lose mass and tone. Intestinal hernias may develop if intestines can bulge out through a weakened abdominal wall.

Birth canal (in women): Changes in pelvic shape in response to bipedalism, together with babies born with larger skulls, can cause childbirth problems if medical intervention is not possible.

Skin cancer: With the loss of shielding body hair, the skin is exposed to larger amounts of UV radiation from the sun.

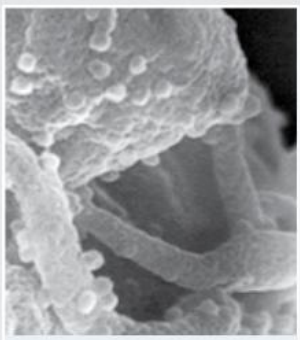
Weakened bones: Bones demineralize with age, as a result they can break more easily.

Varicose veins: An upright posture hampers venous return, allowing blood to collect in the leg veins.

Joint wear: Joint lubricants become thin through repeated use. Joints grind against each other and become damaged.

New Selection Pressures

- Pollution introduces toxins into the environment. Children are particularly vulnerable to environmental toxins. An ability to tolerate and metabolise toxins may provide an evolutionary advantage.
- Resistance to malaria and HIV/AIDS will be strongly favoured in many of the world's most populous regions where there is a high prevalence of these diseases.
- Global dietary changes provide strong selection pressure for food tolerances, e.g. the gene associated with lactose tolerance has been shown to be strongly favoured.



HIV virions budding from a human lymphocyte

CDC

The Consequences

The success of humans as a species has presented modern populations with a number of complex problems. The fast-paced development of new technologies has contributed to global pollution and resource depletion. Medical advances and improved living standards have increased the human life-span, particularly in developed countries, and the world now faces over-population. The ability to manipulate the genetic composition of organisms has many biological, social, and ethical implications that often divide both the scientific community and the general public.

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LINK 214 LINK 84 LINK 82 KNOW

1. Describe the evolutionary trends associated with the following human anatomical features:

   (a) Body hair: _____

   _____

   (b) Jaw: _____

   _____

   (c) Brain: _____

   _____

   (d) Pelvis: _____

   _____

   (e) Face: _____

   _____

   (f) Femur: _____

   _____

2. Describe three problems with the modern human anatomy that have developed as a result of bipedalism:

   (a) _____

   _____

   (b) _____

   _____

   (c) _____

   _____

3. Describe a problem that has resulted from the jaw becoming smaller: _____

   _____

   _____

4. Describe two problems that have arisen because of the reduction of coarseness of body hair:

   (a) _____

   _____

   (b) _____

   _____

5. Modern humans face selection pressures quite different from that of our early ancestors. Describe some of the new selection pressures that operating on the human species:

   _____

   _____

   _____

6. Describe an example of a modern technology or practice and discuss how it might influence future human populations:

   _____

   _____

   _____

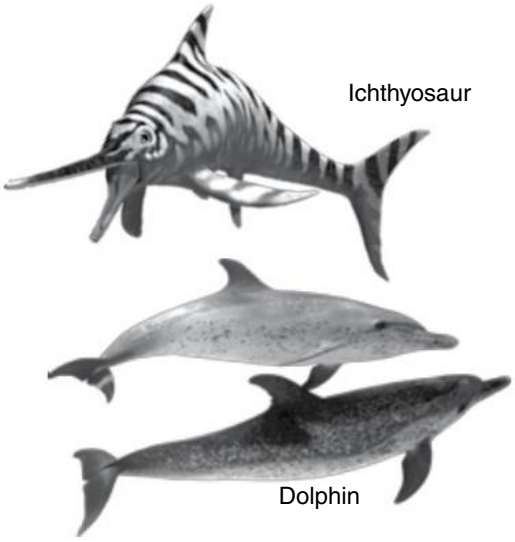
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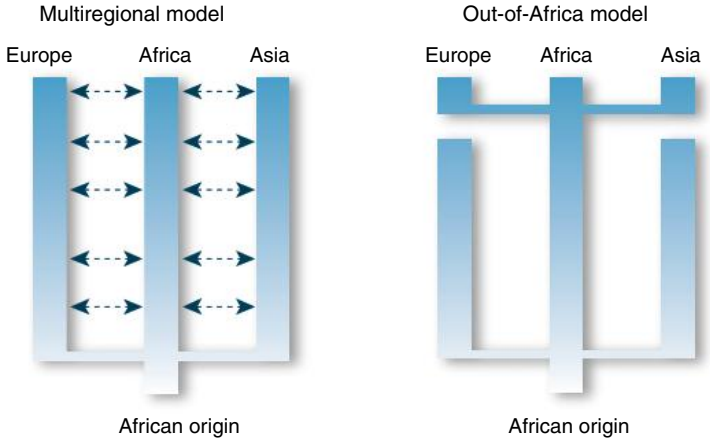
283 Change Over Time Synoptic Question

1 Large marine reptiles known as ichthyosaurs were common in ancient Australian waters. Their outward appearance is similar to the dolphins that inhabit Australian waters today, and they probably behaved in very much the same way. Evidence suggests that at least some species of ichthyosaur gave birth to live young at sea, as dolphins do now.

Discuss the pattern of evolution illustrated by this example. In your answer you should consider the selection pressures acting on ichthyosaurs and dolphins during their evolution. You may use more paper if required.



2. Two important models for human dispersal are the out-of-Africa model and the multiregional model.



Evidence from stone tools suggests *Homo erectus* reached China 1.27 million years ago. The most recent fossil finds suggest it remained there until 143,000 years ago.

Analysis of mitochondrial DNA shows there are 6 different groups within the human population. Only one of these groups, L3, is found outside of Africa. L3 is believed to have arisen about 100,000 years ago.

Analysis of DNA from the Y chromosome indicates that all living males today are related to a man that lived between 140,000 and 500,000 years ago, probably in Africa.

Discuss which of the models for human dispersal (shown above) the data support. In your answer you should:
- describe the out-of-Africa and muliregional models
- explain what the data above suggest about the dispersal of the genus *Homo*
- discuss how the data support (or do not support) the models above

You may use extra paper if required.

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Appendix

Questioning terms in biology

The following terms are often used when asking questions in examinations and assessments.

Analyse:	Interpret data to reach stated conclusions.
Annotate:	Add brief notes to a diagram, drawing or graph.
Apply:	Use an idea, equation, principle, theory, or law in a new situation.
Calculate:	Find an answer using mathematical methods. Show the working unless instructed not to.
Compare:	Give an account of similarities between two or more items, referring to both (or all) of them throughout.
Construct:	Represent or develop in graphical form.
Contrast:	Show differences. Set in opposition.
Define:	Give the precise meaning of a word or phrase as concisely as possible.
Derive:	Manipulate a mathematical equation to give a new equation or result.
Describe:	Define, name, draw annotated diagrams, give characteristics of, or an account of.
Design:	Produce a plan, object, simulation or model.
Determine:	Find the only possible answer.
Discuss:	Show understanding by linking ideas. Where necessary, justify, relate, evaluate, compare and contrast, or analyse.
Distinguish:	Give the difference(s) between two or more items.
Draw:	Represent by means of pencil lines. Add labels unless told not to do so.
Estimate:	Find an approximate value for an unknown quantity, based on the information provided and application of scientific knowledge.
Evaluate:	Assess the implications and limitations.
Explain:	Provide a reason as to how or why something occurs.
Identify:	Find an answer from a number of possibilities.
Illustrate:	Give concrete examples. Explain clearly by using comparisons or examples.
Interpret:	Comment upon, give examples, describe relationships. Describe, then evaluate.
List:	Give a sequence of answers with no elaboration.
Measure:	Find a value for a quantity.
Outline:	Give a brief account or summary. Include essential information only.
Predict:	Give an expected result.
Solve:	Obtain an answer using numerical methods.
State:	Give a specific name, value, or other answer. No supporting argument or calculation is necessary.
Suggest:	Propose a hypothesis or other possible explanation.
Summarise:	Give a brief, condensed account. Include conclusions and avoid unnecessary details.

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The writing team would like to thank the following individuals and institutions who kindly provided photographs, illustrations, or information for this edition:

• Dr Caceci, VA Maryland Regional Veterinary College of Medicine • Dartmouth College and Dr Louisa Howard for TEMs of cell structures • Ed Uthman for the image of the nine week human embryo • Cereal Research Centre, Agriculture and Agri-Food Canada, for their photograph of a pheromone trap • D. Fankhauser, University of Cincinnati, Clermont College for his photograph of Pacinian corpuscles, • Roger LeMoyne UNICEF (www.unicef.org.nz) for the photo of the child taking ORS • Charles Goldberg, University of California, San Diego School of Medicine, for the photograph of a patient with rheumatoid arthritis • Dr. Nita Scobie, Cytogenetics Department, Waikato Hospital for chromosome photos • Wadsworth Centre (NYSDH) for the photo of the cell undergoing cytokinesis • • Genesis Research and Development Corp. Auckland, for the photo used on the HGP activity • Greenpeace for photos used for GMO ethics • Andrew Dunn (www.andrewdunnphoto.com • Rita Willaert for the photograph of the Nuba woman • Aptychus for the photo of the Tamil girl • Dept. of Natural Resources, Illinois, for the photograph of the threatened prairie chicken • Rocky Mountain Laboratories, NIAID, NIH • Aviceda for photos of treecreepers • David Wells(AgResearch) for his photos of cloning • Edward L. Barnard, Florida Department of Agriculture and Consumer Services, Bugwood.org for the photo of the phytotphthora stunted pine • John H. Ghent, USDA Forest Service, Bugwood.org for the photo of phytophthora root rot • Wintec

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Contributors identified by coded credits are as follows: BF: Brian Finerran (Uni. of Canterbury), BOB: Barry O'Brien (Uni. of Waikato), CDC: Centers for Disease Control and Prevention, Atlanta, USA, DH, Don Horne, EII: Education Interactive Imaging, HGSI: Dena Borchardt at Human Genome Sciences Inc. LBS: Lissa Bainbridge-Smith MPI: Max Planck Institute for Developmental Biology, Germany, NASA: National Aeronautics and Space Administration, NCI: National Cancer Institute, NIH: US National Institute of Health, RA: Richard Allan, RCN: Ralph Cocklin, USDA: US Department of Agriculture, WMU: Waikato Microscope Unit.

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