



Science
Schools

Biology



GRADE
10



Express Publishing

Biology

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10

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Introduction

For the student

Welcome to your new Biology textbook, *Grade 10 Biology*. Your textbook comes with a **Grade 10 Biology Student's Portfolio** and a range of *digital resources*. As well as deepening your understanding of key areas of Biology, this book aims to develop your learning skills in science. You will develop these skills in class, in laboratory practicals and whilst conducting research within and outside of class with your fellow students. An emphasis will be placed throughout this course on your ability to present core concepts, research and data effectively to others.

Glossary

A comprehensive glossary is included at the back of this book.

For the teacher

Written for the new Grade 10 Biology subject programme in Kazakhstan, *Grade 10 Biology* aims to meet the broad range of learning objectives set out in the Grade 10-11 Biology subject programme document. It focuses on developing learners' knowledge of and about science through the four content and skill strands outlined in the subject programme:

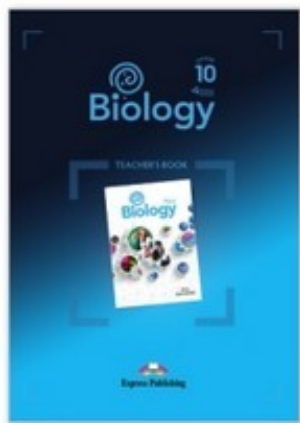
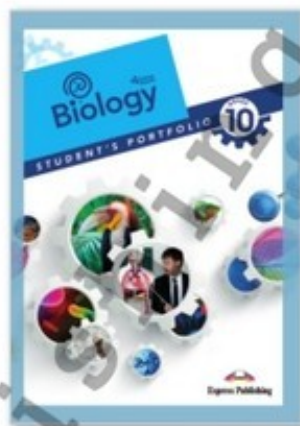
- Understanding of core subject areas in Biology
- Research and experimentation in science
- Communication in science
- Science and society

Key features of the textbook

- **Learning outcomes** are clearly stated at the beginning of each module in student-friendly language
- **Activities** and practical demonstrations allow students to build on their knowledge through guided observation, laboratory practicals and research
- **Diagrams** have been fully labelled and are drawn in a simple style so that learners can replicate them easily
- **Questions** are interspersed within sections of the text to offer teachers the opportunity to use a range of teaching strategies. There are regular opportunities for learners to engage in group work and pair work, discussion, giving of presentations and online research.

Student's Portfolio

The student's Portfolio provides additional revision material and further tasks. The Portfolio enables learners to maintain a detailed record of laboratory practicals, giving them space to reflect on the processes and results of their work. In line with the textbook, it provides detailed sample workings of all stoichiometric calculations they are required to make.



Teacher's Book

A Teacher's Book with full answers to all questions in both the Textbook and Student's Portfolio is provided.

Digital resources

Grade 10 Biology **digital resources** for teachers will further enhance classroom learning. These resources work in conjunction with the Textbook and Student's Portfolio. The resources have been designed to fully integrate with the Textbook to compliment lesson content. Following the principles of the new national Biology subject programme, material is provided to suit a range of learner types and to encourage participation and engagement on the part of the learner.

A series of **videos** allow students to observe science in action across all modules. These videos will reinforce the topic at hand, promote discussion about scientific issues in society and enable teachers to bring a range of perspectives on topics in Biology into the classroom.

Further classroom discussion and participation is opened up through **PowerPoint presentations**, including a thematic presentation of information from the Textbook.

Experiment videos allow for a visual review of laboratory activities and can be used for demonstration or summative plenary work.

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Module 1 Biomolecules and Enzymes

Learning objectives

- Explain the fundamental significance of water to life on Earth (10.4.1.1)
- Classify carbohydrates according to their structure, composition and functions (10.4.1.2)
- Identify reducing and non-reducing sugars (10.4.1.3)
- Describe the chemical structure and function of lipids (10.4.1.4)
- Classify proteins by their structure, composition and functions (10.4.1.5)
- Investigate the influence of various conditions on the structure of proteins (10.4.1.6)
- Test for the presence of proteins (10.4.1.7)
- Investigate the effects of various conditions on enzyme activity (10.1.2.1)

The need for food

Nutrition is the way in which an organism obtains and uses food. Nutrients are the chemical substances, present in food, that are used by organisms. Nutrients are essential to maintain metabolism and continuity of life for all living organisms. In particular, nutrients are necessary:

- As a source of energy
- To make chemicals needed for metabolic reactions
- As the raw materials for the growth and repair of structures in the organism.

The elements present in food

Food is mainly made of 14 elements. Apart from carbon, hydrogen and oxygen, the rest of these elements are often called minerals.

- The six common elements found in food are: carbon (C), hydrogen (H), oxygen (O), nitrogen (N), phosphorus (P) and sulfur (S). The first four of these elements make up over 99% of the mass and atoms present in living organisms. Most of the chemical compounds found in living things are made from carbon atoms bonded together. Compounds made from carbon are said to be **organic** compounds.
- Five elements that are present as dissolved salts are: sodium (Na), magnesium (Mg), chlorine (Cl), potassium (K) and calcium (Ca).
- The three trace elements are: iron (Fe), copper (Cu) and zinc (Zn). Trace elements are elements that are only required in tiny amounts in the diet.



Biomolecules are chemicals that are made inside a living thing.

Biomolecules

Biomolecules contain carbon and are also called biochemicals. The four major types of biomolecules found in food are: carbohydrates, lipids (fats, oils), proteins and vitamins.

Carbohydrates

Elements in carbohydrates

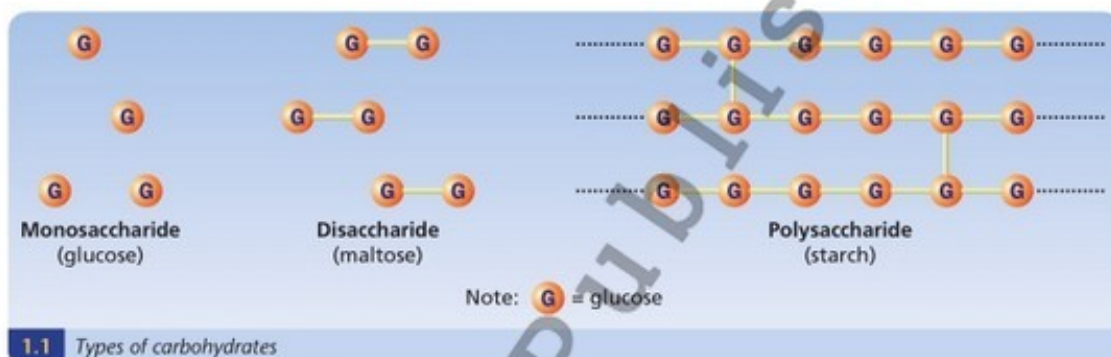
The elements present in carbohydrates are indicated by the name itself: carbon (C), hydrogen (H) and oxygen (O).

These elements are usually present in the ratio $C_x(H_2O)_y$, where x and y are the same number (i.e. $x = y$). This means there is twice as much hydrogen as carbon or oxygen in a carbohydrate.

Glucose is a simple carbohydrate in which x and y are both equal to 6. The formula for glucose is $C_6H_{12}O_6$.

Types of carbohydrates

There are three types of carbohydrates: monosaccharides, disaccharides and polysaccharides.



Monosaccharides

Monosaccharides are carbohydrates composed of a single sugar unit. A single sugar unit is a ring of carbon atoms. Monosaccharides are the simplest and smallest type of carbohydrate. They are sweet to taste and are soluble in water.

Glucose and fructose are examples of monosaccharides.

- Glucose is a very common molecule in biology. It is made by plants in photosynthesis and is the main molecule from which living things get their energy. It is commonly found in sweets, chocolate, fruit and soft drinks.
- Fructose has the same formula as glucose (however, its atoms are arranged differently). It is common in fruits and is much sweeter than glucose.

Disaccharides

Disaccharides are carbohydrates composed of two sugar units joined together. Like monosaccharides, they are sweet-tasting and soluble in water. Examples include maltose and sucrose.

- Maltose is found in germinating seeds and is composed of two glucose molecules joined together.
- Sucrose, or table sugar, is composed of a glucose joined to a fructose.

Polysaccharides

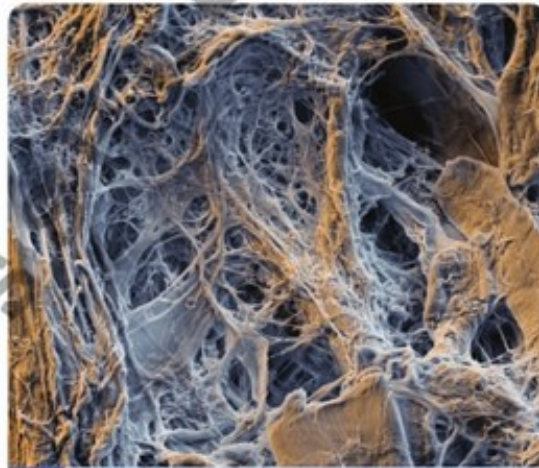
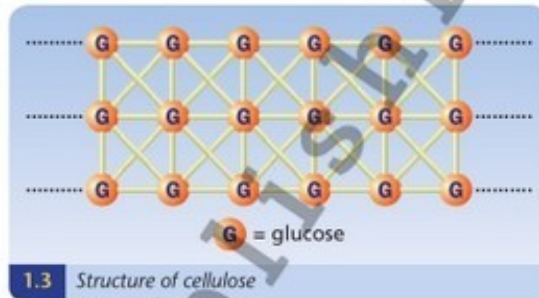
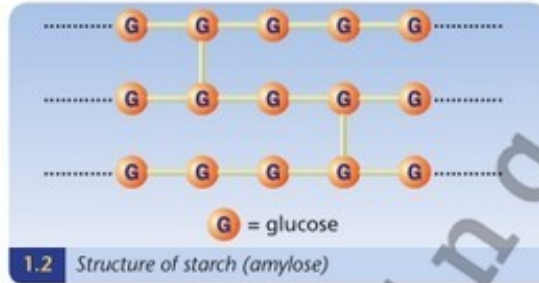
Polysaccharides are carbohydrates composed of many sugar units.

Polysaccharides are insoluble or only slightly soluble in water and are not sweet-tasting. They are very large molecules, often consisting of thousands of monosaccharides. Examples include starch, cellulose and glycogen.



Note that all monosaccharides and some disaccharides (e.g. maltose, but not sucrose) are reducing sugars.

- Starch (also called amylose) is made of many glucose molecules joined together. It is the carbohydrate stored by plants. Common examples of starch are bread, potatoes, rice and pasta. Starch is easily digested as the glucose molecules are arranged in a chain. To extract a glucose it is only necessary to break two bonds (see diagram 1.2).
- Cellulose is also composed of many glucose molecules linked together. However, in cellulose there is much more cross-bonding than there is in starch (see diagram 1.3). This cross-bonding also means that cellulose is:
 - ▶ Very strong (this is why it is used in the structure of cell walls)
 - ▶ Very difficult to digest (we use it as fibre or roughage in our diet).
- Glycogen is a complex polysaccharide. It is composed of large numbers of glucose molecules arranged in many-branched chains. Animals store glycogen in their liver and muscles.



Sources of carbohydrate

Common sources of carbohydrate in our diet are bread, potatoes, rice, pasta, sugars, fruits, sweets and cakes.

Structural role of carbohydrates

Cellulose is used to form plant cell walls.



Metabolic role of carbohydrates

- Glucose is broken down in respiration to release energy. This energy is used to carry out many other metabolic reactions.
- Glucose is made in photosynthesis.

Lipids (fats and oils)

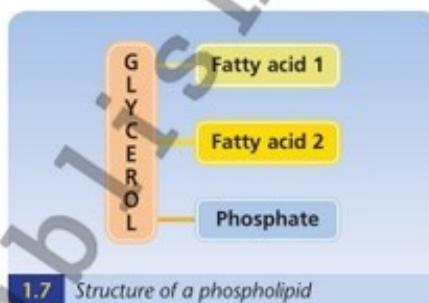
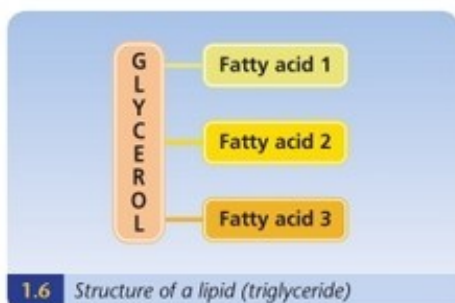
Elements in lipids

Lipids contain the elements carbon, hydrogen and oxygen. Unlike carbohydrates, the elements in lipids have no simple ratio. However, lipids have very little oxygen.

Fats are lipids that are solid at room temperature (20°C). Oils are lipids that are liquid at room temperature.

Structure of lipids

The smallest lipids are made of one molecule of glycerol linked to three fatty acid molecules. This structure is called a triglyceride. Different fats and oils have different types of fatty acids.



Phospholipids are important in the structure of cell membranes.

Sources of lipids

Sources of lipids in our diet are butter, oils, margarine, cream, fat on meat and fried food. Lipids stain clothing.

Structural role of lipids

- Lipids are important food (or energy) stores in plants and animals. One gram of lipid contains twice as much energy as a gram of carbohydrate. This means that lipids can store twice as much energy compared with an equivalent amount of carbohydrate. This is especially important for animals that have to carry their stored energy around with them. In animals, the stored lipids have secondary functions, such as heat insulation (fat under the skin) and protection of organs (fat around the heart and kidneys).
- Lipids combine with phosphorous to form phospholipids and with proteins to form lipoproteins. Both of these are important in the structure of cell membranes.

Phospholipids are fat-like substances in which one of the fatty acids is replaced by a phosphate group or has a phosphate group added to it.

Metabolic role of lipids

Lipids can be broken down in respiration to release energy.

Proteins

Elements in proteins

Proteins contain the elements carbon, hydrogen, oxygen and nitrogen. They sometimes contain smaller amounts of sulfur and some may contain phosphorus and other elements. There is no ratio for the elements in a protein, but proteins are very large and complex, often containing tens of thousands of atoms.

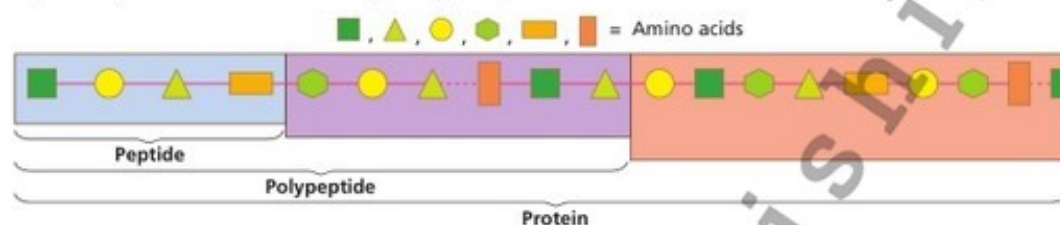
Structure of proteins

Proteins are composed of amino acids. There are 20 common amino acids found in proteins.

The bond between amino acids is called a peptide bond. A peptide is made of a small number of amino acids (less than 20).

A polypeptide has more than 20 amino acids.

A protein is a long polypeptide (at least 200 amino acids). The amino acids that make up a protein can be thought of as the letters in an alphabet. By combining them in different sequences, nature can make a huge range of proteins.



1.8 Relationship between amino acids, peptides, polypeptides and proteins

However, the way in which a protein works does not depend on the amino acid sequence alone. The manner in which the proteins are **folded** to take up three-dimensional (3-D) shapes is equally important.



The importance of proteins folding in the correct way is seen when they fold incorrectly. Prions are proteins that do not fold correctly. They cause similar proteins to fold incorrectly and are responsible for brain and nervous system diseases such as bovine spongiform encephalopathy (BSE; in cattle), Creutzfeldt-Jakob disease (CJD; in humans) and scrapie (in sheep).

Proteins can be classified according to structure, composition and function.

Classification of proteins by structure

Proteins can be **globular** or **fibrous**.

Globular proteins are folded into a ball shape. Their shape determines their function and they are destroyed if their shape is changed. This is called denaturing and is not reversible. Globular proteins are water-soluble and have metabolic roles. Examples include all enzymes, haemoglobin and albumin.

Fibrous proteins are elongated like a fibre or rope. They are not water soluble and generally have structural roles. Examples include keratin (found in hair and nails) and collagen (found in skin and connective tissue).



1.9 Globular protein



1.10 Fibrous protein

Classification of proteins by composition

Some proteins are made from just one chain of amino acids such as lysozyme which is an enzyme that breaks down the cell walls of bacteria.

Other proteins are made from more than one chain, such as collagen (3 long chains twisted together), insulin (2 globular chains) and antibodies (4 or more globular chains).

Some proteins contain parts that are not made from amino acids. These parts are called prosthetic groups. Haemoglobin has a prosthetic group called haem which holds the iron.

Classification of proteins by function

Proteins can have either a **metabolic** function or a **structural** function.

Many metabolic proteins are enzymes, but metabolic proteins also include some hormones like insulin. Metabolic proteins are globular and water soluble. This helps them to be moved easily to where they are needed.

Structural proteins are fibrous and are found in the skeleton, muscles and the tissues that hold organs in place. It is important that they are not water soluble. Just imagine what would happen if your hair dissolved when it got wet!

Sources of protein

Sources of protein include meat, fish, eggs, nuts, milk, peas and beans.

It is important to note that amino acids are not stored in the body. Surplus amino acids are taken to the liver and converted into urea, which is a toxic waste product. This process is called deamination. Urea is carried by the blood from the liver to the kidneys. In the kidneys, urea becomes part of urine and is excreted.

Structural role of proteins

Fibrous protein such as keratin is found in skin and hair. Myosin is found in muscle.

Metabolic role of proteins

Proteins are used as enzymes to control reactions. They also form antibodies to fight infection. Some hormones are protein-based and are used to regulate body reactions.

Energy transfer reactions

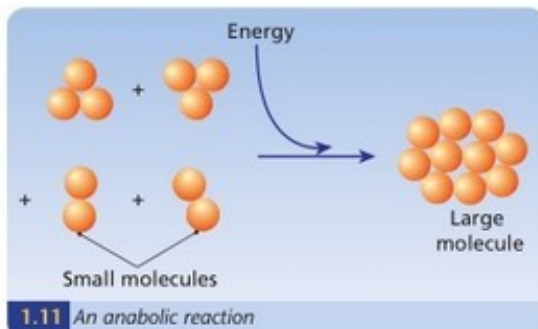
All the reactions taking place in an organism are referred to as its metabolism. Metabolic reactions can be divided into **anabolic reactions** and **catabolic reactions**.

Anabolic reactions



Anabolic reactions use energy to convert smaller molecules into larger molecules.

Examples of anabolic reactions are the formation of muscle from amino acids (also called protein synthesis), the formation of cellulose from glucose and photosynthesis (where carbon dioxide and water are used to make a food such as glucose).



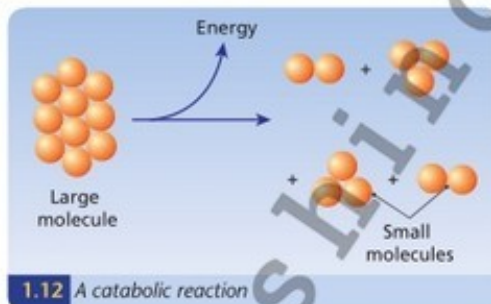
Anabolic steroids are drugs that are used (illegally) in sports to build up muscles.

Catabolic reactions



Catabolic reactions release energy when a complex molecule is broken down to a simpler form.

Catabolic reactions include respiration (in which a molecule of food is broken down to release energy), the digestion of food and the decay of dead plants and animals.



Questions on Module 1

- Give the name and chemical symbol for:
 - The six most important elements in organisms.
 - The five elements found in dissolved salts.
 - Three trace elements.
- Explain what is meant by biomolecules.
 - Name four types of biomolecules found in food.
- What elements are present in:
 - Table sugar (sucrose)
 - Meat
 - Butter?
- Distinguish between monosaccharides, disaccharides and polysaccharides.
 - Name one biomolecule from each of these three categories.
- Name three polysaccharides and give one use for each of them.
- A sample of urine when boiled with Benedict's solution turned red. What does this result tell you: (a) about the urine and (b) about the person from whom it was taken?
- Name the four elements in phospholipids.
 - Give one use for phospholipids.
- State one structural role and one metabolic role for: (a) Carbohydrates (b) Lipids (c) Proteins.
- What is meant by:
 - Metabolism
 - Anabolism
 - Catabolism?
 - State two examples of each process.
- A picnic basket consists of brown bread, butter, apples, oranges, milk, ham, salmon, salt, cakes.
 - Name one good source from this list for:
 - Carbohydrate
 - Protein
 - Fat
 - Vitamin C
 - Vitamin D
 - Roughage
 - Calcium
 - Phosphorus.
 - Name three different carbohydrates and their sources from the above list.
 - Name a water-soluble and water-insoluble (i) carbohydrate and (ii) vitamin found in these foods.
- What is a solvent?
 - Give two biological benefits of water being a good solvent.
- Name three reactions in which water plays a role.
- Say whether the following statements are true or false. In the case of a false statement, give a reason why it is false.
 - Carbohydrates contain hydrogen and oxygen in the same ratio as water (H_2O).
 - Amino acids do not contain nitrogen.
 - Fish is a good source of protein and lipid, but not carbohydrate.
 - Amino acids are required to make fatty acids.
 - Keratin is a protein found in egg white.
 - The Biuret test indicates the presence of proteins.
 - Chewing food is an example of anabolism.

Experiment 1.1 To conduct qualitative tests for food

A qualitative test measures whether a substance is present or absent.

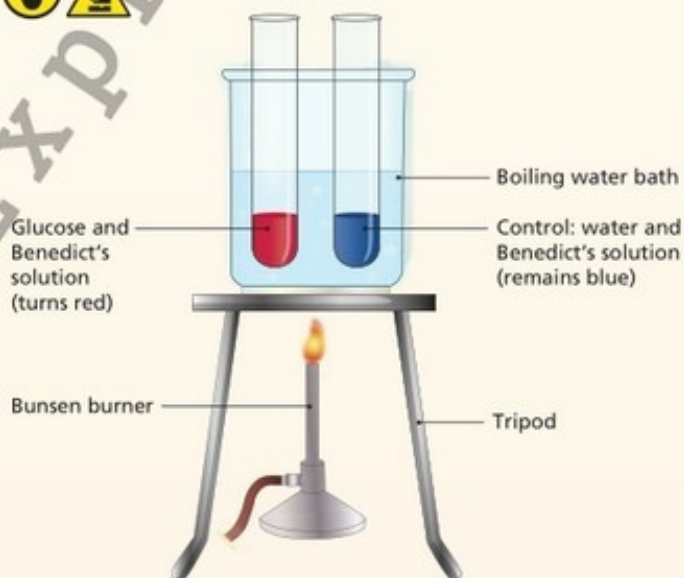
Experiment 1.1a To test for reducing sugar

- 1 Dissolve glucose in water in a test tube.
- 2 Add an equal volume of Benedict's solution (which is blue).
- 3 In a second test tube mix equal volumes of water and Benedict's solution. This will act as a control.
- 4 Heat the test tubes in a boiling water bath.
- 5 If reducing sugar is present, the solution turns red (often called brick red).
- 6 If reducing sugar is not present the solution remains blue.

Note: Fehling's solution can be used instead of Benedict's solution.



1.13 Benedict's test for reducing sugar; a negative result (blue) and a positive result (red-orange)



1.14 Testing for reducing sugar



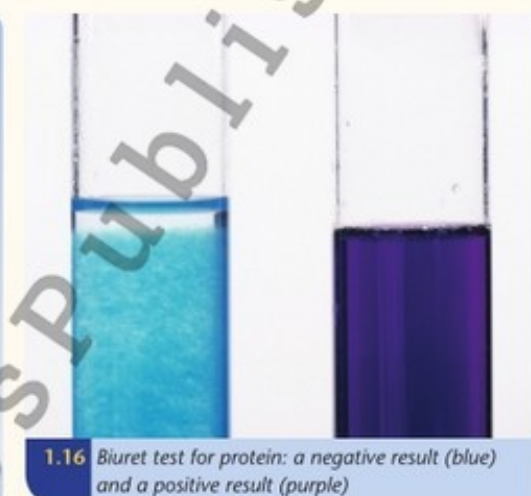
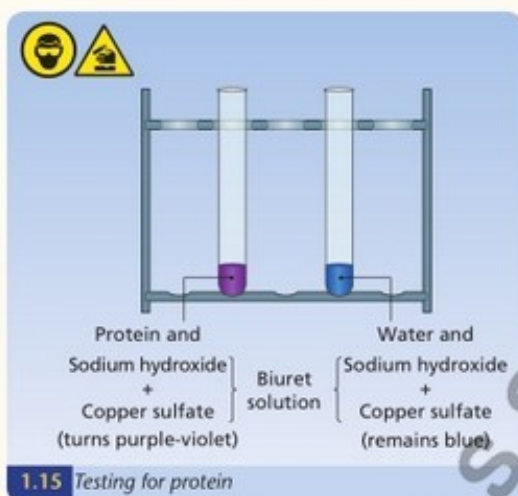
Experiment 1.1b To test for protein

- 1 Dissolve a sample of soluble protein (e.g. egg white or milk) in water.
- 2 Add sodium hydroxide (colourless) until the solution clears.
- 3 Then add a few drops of dilute copper sulfate (blue).

Note: as an alternative to steps 2 and 3, add an equal volume of Biuret solution.

This contains sodium hydroxide and copper sulfate and is blue.

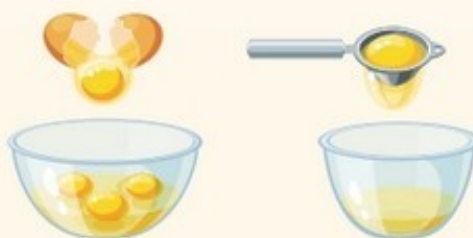
- 4 As a control, add sodium hydroxide and copper sulfate (or Biuret solution) to water.
- 5 The appearance of a purple-violet colour shows that proteins are present.
- 6 If protein is not present the colour remains blue.

**Experiment 1.2 The effect of temperature and pH on protein structure**

The protein present in egg white is albumin which is a globular protein. As its structure changes, its texture and appearance change.

- 1 Take a fresh egg and crack it into a bowl.
- 2 Separate the yolk and white of the egg into two beakers.

Keep the whites of the eggs for the experiment. The yolks are not needed.



A Effect of temperature

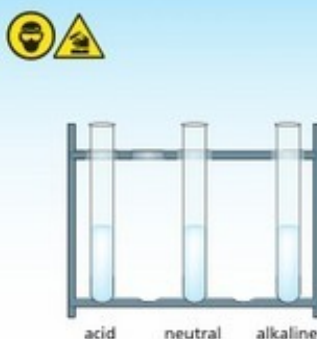
- 1 Pour a small volume of egg white into a test tube. There should be enough to about quarter-fill the test tube.
- 2 Place the test tube with the egg white into a water bath and heat it.
- 3 Record the temperature at which the egg white starts to change texture and appearance.



1.18 Testing for effect of temperature on protein structure

B Effect of pH

- 1 Pour a small volume of egg white into three different test tubes in a rack. There should be enough to about quarter-fill each of the test tubes.
- 2 Label one test tube **acid**, one **neutral** and one **alkaline**.
- 3 Slowly add, drop by drop, an acid to the test tube labelled **acid**, mixing the contents after each drop is added. You can use vinegar or 1 mol/dm³ (1M) hydrochloric acid.
- 4 Record your observations.
- 5 To the test tube labelled **neutral**, slowly add water, drop by drop and record your observations. Mix the contents after each drop as before.
- 6 To the test tube labelled **alkaline**, slowly add 1 mol/dm³ (1M) sodium hydroxide and record your observations. Mix the contents after each drop as before.



1.19 Testing for effect of pH on protein structure

Metabolism

Metabolism is the sum of all the chemical reactions that take place within an organism. These reactions involve growth, movement, maintenance of a constant internal state, repair, response to stimuli and reproduction. Each of these changes requires that energy is released or absorbed. This means that metabolism is closely associated with energy conversions.

Metabolism is necessary to control the chemical and energy requirements of a cell. By doing this, metabolism maintains a balanced internal state (called homeostasis) within an organism.

Sources of energy



1.20 Solar panels absorb light energy

Solar energy

The primary source of energy for life on Earth is sunlight. Some of the energy in sunlight (solar energy) is trapped by organisms that contain pigments which can absorb light. Chlorophyll is one of these pigments.

Producers such as green plants use solar energy to form the chemical bonds of carbohydrates and other biomolecules. This form of energy conversion is carried out by the process of photosynthesis.

Cellular energy

Cellular energy refers to sources of energy that are capable of being released by reactions within a cell, i.e. the energy stored in the bonds of biomolecules such as carbohydrates or lipids.

Some of the chemical energy stored in the bonds of biomolecules is transferred to consumers when they eat producers. The energy can then pass along the entire food chain.

Each organism breaks down energy-rich biomolecules in the process of respiration. This releases energy, some of which is used by the cells, while the rest is released into the environment as heat.

Solar energy is energy from the Sun. Cellular energy is the energy stored in the bonds of biomolecules.

Water

Water is the most abundant chemical in living things. It accounts for 99% of all the molecules in the human body. It comprises 60% of human body mass and 90% of the mass of most plants.

Life originated in water and living things are still dependent on water for their survival. Water is essential to life for three main reasons.

- 1 It is the liquid in which all metabolic reactions take place.
- 2 It provides the basis for transport systems in organisms.
- 3 It is the environment in which many organisms live.

Importance of water for living things

Component of cytoplasm and body fluids

Water is the most common chemical in cells. It is mainly found in the cytoplasm, which is the liquid that surrounds the nucleus in a cell.

In humans, about one-third of the body's water is found outside the cells. Some of this is in the form of tissue fluid, which surrounds all body cells, and the rest forms plasma, the liquid part of blood.

Good solvent

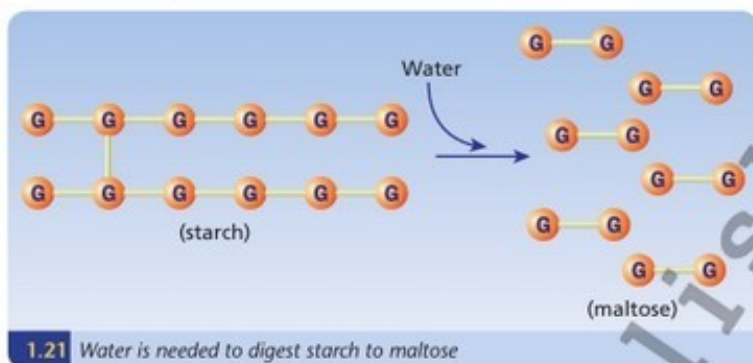
Water is a good solvent, i.e. it is able to dissolve a wide range of molecules.

- This allows chemical reactions to take place in water in the cytoplasm and in cell organelles.
- It also allows many molecules to be dissolved in water for transport in plants and animals.

Participates in chemical reactions

Water is directly involved in a number of biochemical reactions. These include:

- Photosynthesis: in which water is used to supply hydrogen ions and electrons
- Respiration: in which water is formed as an end product
- Digestion: in which water is needed to break down food. This is why we should take a drink when eating food.



Movement through membranes

Water can easily pass in or out through biological membranes.

When cells absorb large amounts of water they become swollen. If cells lose water they shrivel and lose their shape. The loss of shape of cells can have serious results for the function of the cell. For example, if red blood cells lose shape, they absorb and carry less oxygen. If plant cells lose shape, the overall plant may also lose shape (a process called wilting).



Good absorber of heat

Water is a good absorber of heat energy. This means it is slow to heat up and slow to cool down. As a result:

- The oceans and other large bodies of water (and the organisms in them) have relatively stable temperatures
- The high water content of organisms helps to keep their temperature stable. This allows biological reactions to take place over a narrow temperature range (which means the reactions do not speed up or slow down due to the heating or cooling of the organism).

Enzymes

Enzymes are catalysts made of protein. This means that enzymes are often called biological (or organic) catalysts.

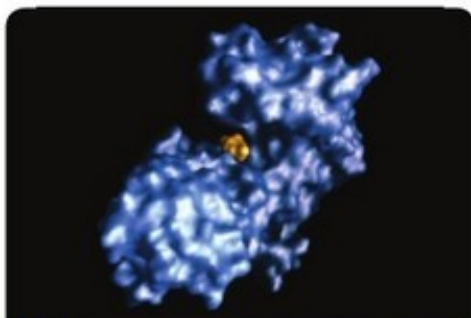
Enzymes are used to speed up chemical reactions and allow them to proceed at normal cell temperatures.



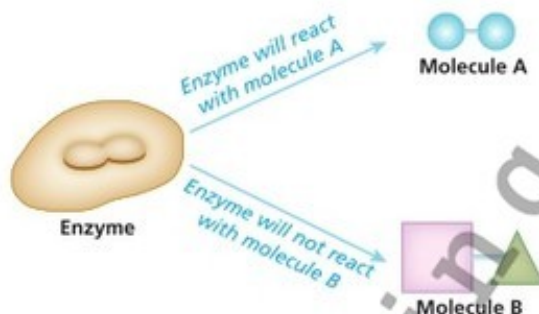
Note that while all enzymes are proteins, not all proteins are enzymes.



A catalyst is a substance that speeds up a reaction, without itself being used up in the reaction.



1.23 An enzyme (blue) with its substrate (brown)

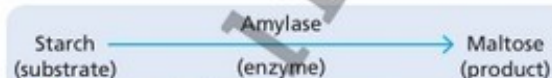


1.24 The importance of enzymes having the right shape

Anything that changes the shape of an enzyme will reduce the efficiency of the enzyme to speed up a reaction. Changing the pH or temperature of a reaction will change the shape of enzymes, which in turn will affect the speed of the reaction.

Features of enzymes

As an example of a typical enzyme reaction consider the reaction in diagram 1.25.



1.25 A sample of an enzyme-controlled reaction

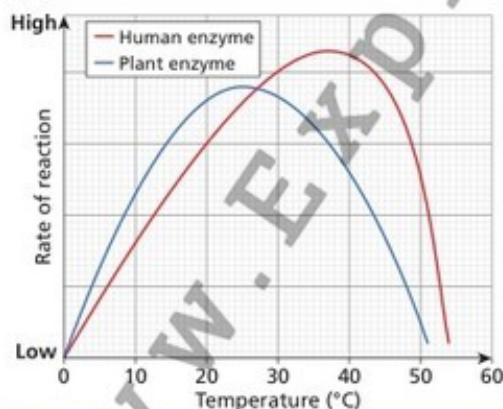
- Enzymes are made of protein.
- Enzymes work because they have the correct **shape** to fit the substrate. This means the enzyme must have a complex, 3-D shape in order for it to fit with the substrate.
- Enzyme reactions are **reversible**. Just as a key can either open or close a lock, an enzyme can cause a reaction to proceed in either direction. This means any enzyme can be anabolic (forms more complex compounds) or catabolic (breaks down larger compounds).

A denatured enzyme has lost its shape and can no longer function.

Factors affecting enzyme activity

Enzymes work best under certain ideal conditions. Any change in these conditions will effect the rate of the reaction. These conditions include temperature and pH.

Enzymes are proteins that speed up a reaction without being used up in the reaction. The **substrate** is the substance with which an enzyme reacts. The **product** is the substance(s) the enzyme forms.



1.26 The effect of temperature on the rate of reaction

Human enzymes work best at 37°C (body temperature), whereas most plant enzymes work best at 20-30°C.

Above a certain temperature, enzymes begin to lose their 3-D shape. As a result the rate of reaction falls. When the shape of an enzyme is fully lost (usually above 50°C), the enzyme is said to be **denatured**. In this condition it has lost its ability to function.

Enzymes may also be denatured by other factors such as unsuitable pH, inhibitors and radiation.

pH

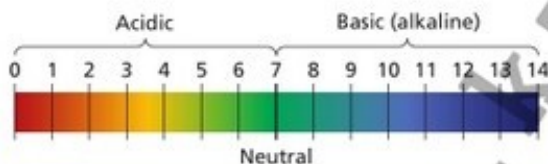
The pH scale runs from 0 to 14. Values

between 0 and 7 are acidic, pH 7 is neutral and values between 7 and 14 are basic (or alkaline).

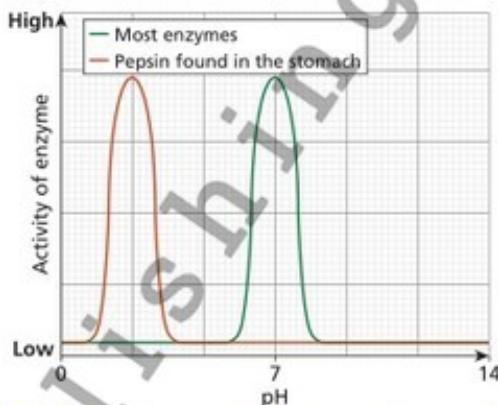
Enzymes are very sensitive to changes in pH. As a result enzymes only work over a very narrow pH range. For most enzymes this is pH 6-8. Outside this range the activity of the enzyme falls quite rapidly. This is because the enzyme loses its shape, i.e. it becomes denatured. The optimum (or ideal) pH for most enzymes is pH 7.

Note: Pepsin, an enzyme in the stomach, works best at a pH of 2. This allows it to work efficiently in the acid conditions of the stomach.

The way in which pH affects the rate of reaction is shown in diagram 1.28.



1.27 The pH scale



1.28 The effect of pH on the rate of reaction



1.29 Bioreactors producing protein drugs

Immobilised enzymes

Traditionally, bioprocessing involved the use of microorganisms, such as yeast and bacteria, to produce foodstuffs such as cheeses, yoghurts, breads, beers and wines.

In recent times, purified enzymes have been used in bioprocessing to produce a vast range of

products, including antibiotics, drugs, vaccines, methane gas (biogas), food colourings and flavours, vitamins, amino acids, sugar syrups, enzymes and perfumes.

To prevent wasting enzymes they are often immobilised or fixed so they remain in place. In this way they remain in the bioreactor and are easier to recover and reuse.

Advantages of immobilised enzymes

- Immobilised enzymes can be reused. This is an important consideration, because the cost of replacing enzymes can be rather high.
- Immobilised enzymes remain in the reaction vessel at the end of the process. This means the product does not need to be separated from the enzyme.
- Very often the process of immobilising an enzyme increases its stability.

Uses of immobilised enzymes

- Glucose isomerase is used to convert glucose to the sweeter-tasting fructose for use in some foods.
- Penicillin acylase is used to convert penicillin into new antibiotics.

Bioprocessing is the use of enzyme-controlled reactions to produce a product. A **bioreactor** is a vessel or container in which living cells or their products are used to make a product.

Enzyme experiments

In experiments measuring the rate of enzyme action (such as the following experiment) there are four factors to be considered: temperature, pH, enzyme concentration and substrate concentration.

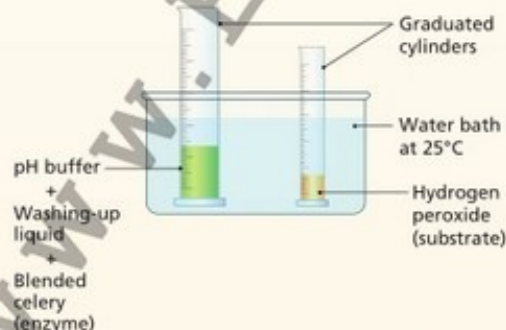
In each of these experiments one factor is varied, while the other three must be kept constant. This is achieved by adopting the following procedures.

Varying the four factors in enzyme experiments		
Factor	Method used to keep factor constant	Method used to vary factor
pH	Use the same pH buffer	Use pH buffers of different values
Enzyme concentration	Add the same volumes of enzyme	Not on the course
Substrate concentration	Add equal volumes of the same substrate	Not on the course
Temperature	Use waterbath(s) at the same temperature	Use waterbath(s) at different temperatures

Experiment 1.3 To investigate the effect of pH on the rate of enzyme activity



1.30 Foam produced when hydrogen peroxide is broken down by catalase (in pieces of potato)



1.31 Investigating the effect of pH on enzyme activity

Introduction

Catalase is an enzyme that is found in a wide range of living things, e.g. liver, radishes, celery and potatoes. It converts the toxic substance hydrogen peroxide (H_2O_2) into water and oxygen.

When using catalase the oxygen forms foam (in association with washing-up liquid). The volume of the foam indicates the activity of the enzyme.

- 1 Place some pH buffer solution 4 in a graduated cylinder (pH buffer 4 ensures that the pH will remain at 4).
- 2 Using a dropper add one drop of washing-up liquid to the graduated cylinder (the washing-up liquid traps the oxygen that is released, forming foam).
- 3 Blend some stalks of celery in water in a blender. Filter this solution into a large beaker using coffee filter paper (filtration removes the blended cells and contents; coffee filter paper allows for fast filtration).

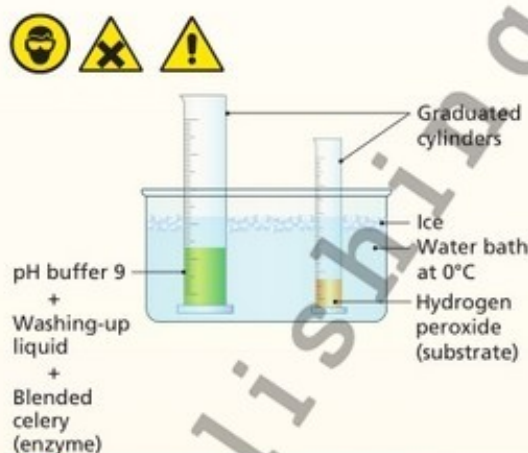
- 4 Add some of the filtrate to the graduated cylinder (the celery contains the enzyme catalase).
- 5 Add some hydrogen peroxide to a smaller graduated cylinder (hydrogen peroxide is the substrate).
- 6 Stand both graduated cylinders in a water bath (or a beaker of water) at 25°C for a few minutes (this ensures a constant temperature).
- 7 Remove the graduated cylinders from the water bath and pour the hydrogen peroxide into the graduated cylinder containing the blended celery.
- 8 Note and record the volume at the top of the foam after 2 minutes.
- 9 Calculate the volume of foam produced. This is done by subtracting the original volume of liquid in the graduated cylinder from the volume at the top of the foam after 2 minutes (the volume of foam indicates the rate of the reaction).
- 10 Repeat steps 1-9 using pH buffers 7, 10 and 13.
- 11 Record the results as shown overleaf; the first set of figures is filled in as an example.

Experiment 1.3 results				
pH buffer	4	7	10	13
Original volume (cm ³)	25			
Volume after 2 minutes (cm ³)	25			
Volume of foam (cm ³)	0			

- 12 Draw a graph of the results. Put pH on the horizontal axis and the volume of foam produced on the vertical axis. The graph should have a similar shape to that in diagram 1.23.
- 13 Note that catalase is different to most enzymes as it has its maximum activity at pH 9 or 10.
- 14 As controls, repeat each procedure but do not add blended celery (i.e. no catalase is present) or add blended celery that has been boiled (to denature the catalase). In each case no foam is formed.

Experiment 1.4 To investigate the effect of temperature on the rate of enzyme activity

- Place some pH buffer 9 solution in a graduated cylinder (catalase works best at pH 9; the buffer ensures the pH remains constant at 9).
- Using a dropper add one drop of washing-up liquid to the graduated cylinder (the washing-up liquid traps the oxygen that is released, forming foam).
- Blend some stalks of celery in water in a blender. Filter this solution into a large beaker using coffee filter paper (filtration removes the blended cells and contents; coffee filter paper allows for fast filtration).
- Add some of this solution to the graduated cylinder (the celery contains the enzyme catalase).
- Add some hydrogen peroxide to a smaller graduated cylinder (hydrogen peroxide is the substrate).
- Stand the graduated cylinders in a large beaker of ice-cold water until they are at 0°C.
- Remove the graduated cylinders from the water bath and pour the hydrogen peroxide into the graduated cylinder containing the blended celery.
- Note and record the volume at the top of the foam after 2 minutes.
- Calculate the volume of foam produced, as shown in diagram 1.32 in Experiment 1.2.
- Repeat steps 1–9 at 10°C, 20°C, 30°C, 40°C, 50°C and 60°C.
- Record the results as shown; the first set of figures is given as an example.



1.32 Investigating the effect of temperature on enzyme activity

Experiment 1.4 results

Temperature (°C)	0	10	20	30	40	50	60
Original volume (cm ³)	25						
Volume after 2 minutes (cm ³)	25						
Volume of foam (cm ³)	0						

- Draw a graph of the results. Put temperature on the horizontal axis and the volume of foam produced on the vertical axis. The graph should have a similar shape to those in diagram 1.21.
- As controls, repeat each procedure but do not add blended celery (i.e. no catalase is present) or add blended celery that has been boiled (to denature the catalase). In each case no foam is formed.

Questions on Module 1

- 14 Distinguish between solar and cellular energy.
- 15 Name the biological process that converts: (a) Solar energy to cellular energy (b) Cellular energy to energy that can be used in a cell.
- 16 Distinguish between: (a) An enzyme and a catalyst (b) A substrate and a product.
- 17 (a) Explain why the shape of an enzyme is important.
 (b) Explain why high temperatures may prevent an enzyme from working.
 (c) Name another factor that can affect the rate of enzyme action.
- 18 Name and give the function of (a) one anabolic and (b) one catabolic enzyme.
- 19 Give a reason for each of the following:
 (a) Amylase is inactivated in the mouth by very hot drinks.
 (b) Amylase works in the mouth but not in the stomach.
 (c) Pepsin works in the stomach, but amylase does not.
 (d) Putting food in a refrigerator slows bacterial action.
 (e) Putting food in a freezer slows bacterial action.
- 20 (a) Explain what is meant by:
 (i) Bioprocessing
 (ii) Immobilised enzymes
 (iii) A bioreactor.
 (b) Give two traditional (or older) and two modern examples of bioprocessing.
 (c) Suggest one problem with the use of freely dissolved enzymes in bioprocessing.
- 21 Give three benefits for using immobilised (compared to free) enzymes.
- 22 Give two uses for immobilised enzymes. In each case name the substrate, enzyme and product of the reaction.
- 23 In testing the effect of pH on enzyme activity:
 (a) How would you keep the temperature constant?
 (b) How could you change the pH?
 (c) Describe a suitable control.
 (d) (i) What result would you expect for the control?
 (ii) If the enzyme was pepsin, at what pH would you expect it to be most effective?
- 24 Answer the following questions with reference to investigating the effect of temperature on enzyme action.
 (a) Name the enzyme and substrate used.
 (b) Why is washing-up liquid used?
 (c) Why are the ingredients placed in a water bath before mixing them?
 (d) What factors were kept constant during this experiment?
 (e) Describe how the factors named in part (d) are kept constant.
 (f) What control should be used in the experiment?
 (g) How is the activity of the enzyme measured?
 (h) (i) Explain why less foam forms at 10°C compared with 20°C.
 (ii) Suggest the pH at which pepsin would be most active.
- 25 Describe the similarities and differences between a triglyceride and a phospholipid.
- 26 Globular proteins are water soluble. Explain why it is essential that fibrous proteins are not water soluble.

Module 2 DNA and RNA

Learning objectives

- To understand the structure and function of DNA [10.4.1.8](#)
- To describe the process of DNA replication based on Chargaff's rules [10.4.1.9](#)
- To understand the structure and functions of RNA types [10.4.1.10](#)
- To compare the structure of DNA and RNA molecules [10.4.1.11](#)

Heredity

Heredity is the passing on of features from parents to offspring by means of genes.

Heredity is also called **genetic inheritance**.

- Humans inherit features such as the number of fingers, the production of nails and the ability to form tears.
- Plants inherit features such as the number of petals, the colour of the petals and the shape of the leaves.

Genes

A gene is a section of DNA that causes the production of a protein.

Many of the proteins produced by genes are enzymes. Genes are said to control a cell because many of the enzymes they produce control cell activities. Genes are the units (or structures) of heredity.

Gene expression

Gene expression is the way in which the genetic information in a gene is decoded in the cell and used to make a protein.

In other words, gene expression refers to the way in which genes work. It describes the sequence of events that occur so that a gene on a chromosome in the nucleus can cause the production of, for example, an enzyme in the cytoplasm of the same cell.

It is the expression of genes that produces the characteristics or traits that are inherited. Characteristics such as those listed earlier arise from the interaction between the genes that are inherited (heredity) and the environment.

- A child may inherit genes for tallness, for instance, but if the child's diet lacks the correct nutrients, the genes may not be able to cause tallness, i.e. the genes may not be expressed.
- In the same way, leaf cells have genes to control the production of the green pigment, chlorophyll. However, if the plant grows in a dark place these genes do not work and chlorophyll is not made.

Characteristics are traits or features that are inherited genetically.

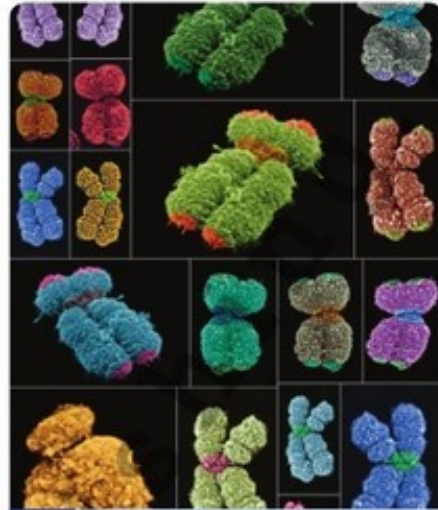
Characteristics arise from the interaction of heredity and the environment.

Chromosomes

Chromosomes are composed of about 60% protein and 40% DNA.

The protein is responsible for holding the DNA in a tightly packed configuration so that it can fit into the nucleus. For example, a typical human chromosome has a DNA strand that could extend to about 6 cm long. This is far too large to fit into a nucleus that is much smaller than the full stop at the end of this sentence.

To enable DNA to fit into a nucleus, it is heavily coiled and folded, very similar to an elastic band twisted repeatedly until it forms a solid ball. Proteins are responsible for holding the DNA in its folded state.

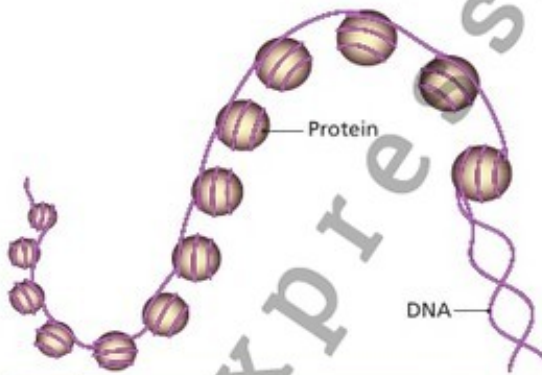


2.1 A variety of chromosomes

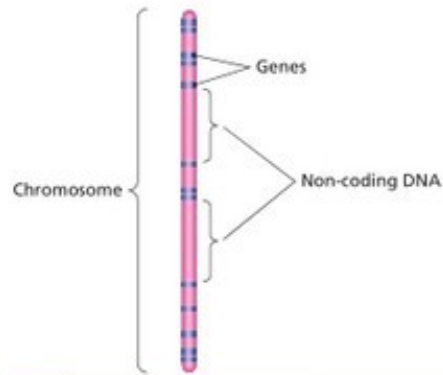
Non-coding DNA

Genes are arranged along the DNA of a chromosome in a line. Sometimes a number of genes are located close together on the chromosome. Other genes are widely separated along the chromosome.

This means that large sections of the chromosome are not made up of working genes. In fact it is known that about 97% of the DNA in a human cell does not consist of genes. This DNA is said to be non-coding (i.e. it does not carry the code for the formation of a protein).



2.2 The structure of a chromosome



2.3 Genes on a chromosome

The non-coding DNA was often called **junk DNA**. Recent research indicates that large amounts of this DNA may act as a genetic control panel switching genes on and off.

Non-coding DNA is of two types:

- Some of it occurs **between** genes
- Some of it is found **within** genes.

The sequence of non-coding DNA varies greatly from one person to another. It is the DNA in these non-coding sections that is used to prepare DNA profiles.

Non-coding DNA is DNA that does not cause the production of a protein.

Structure of DNA (deoxyribonucleic acid)

There are only four different chemicals, called 'bases', used in DNA. The four bases are known by the first letter of their names:

- Adenine (A)
- Thymine (T)
- Guanine (G)
- Cytosine (C).

Each of the four bases can only join or bond with one other base:

- A joins with T
- G joins with C.

The pairs of bases, A/T and G/C, are said to be complementary base pairs.

The DNA molecule is made of two attached strands, similar to the two sides of a ladder (as shown in diagram 2.6). The strands are held together by the complementary bases.

The pattern of complementary base pairing means that if one strand of a DNA molecule has the sequence TAGCAT, then the sequence on the partner strand must be ATCGTA.

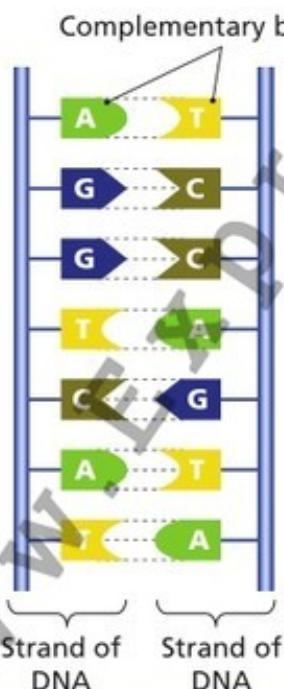
The double-stranded DNA is twisted to form a spiral structure, with each of the side strands forming a spiral or helix. DNA is arranged in this way to form a double helix shape.



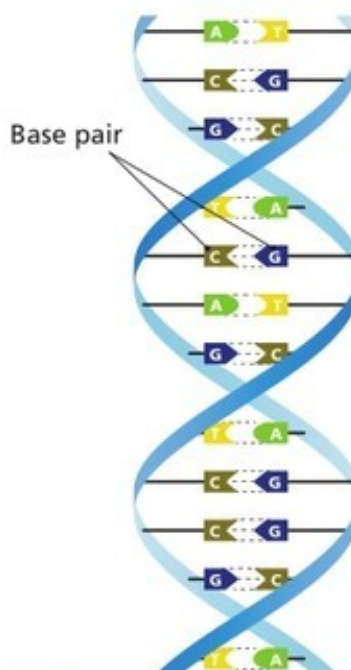
2.4 DNA



2.5 Complementary base pairs



2.6 Simplified structure of DNA



2.7 DNA double helix

The genetic code

A chromosome consists of many base pairs arranged into a double helix. For example, the DNA in the longest human chromosome (number 1) contains about 300 million base pairs. A gene is a sequence of many bases. The precise sequence of bases is called the genetic code.

A gene works, or is expressed, when this code is sent into the cytoplasm (using another molecule called RNA) to form a protein.

Genes are made of DNA. A gene is a section of DNA that instructs a cell to form a particular protein molecule.

Proteins are made up of combinations of hundreds or thousands of amino acids joined together in a specific sequence. Up to 20 different types of amino acids are used in proteins. This means that a gene must carry a different code to control the assembly of each of the 20 different amino acids.

DNA codes for each amino acid by using a sequence of three consecutive bases. Such a sequence of three bases is called a **triplet** (or **codon**).

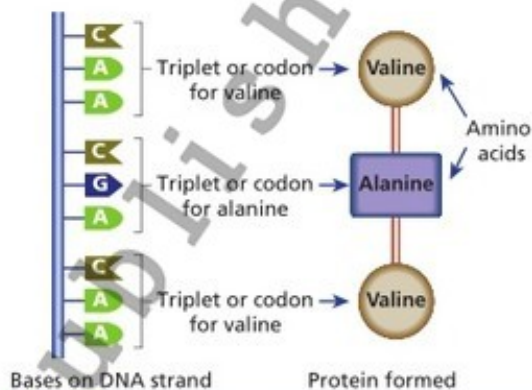
For example, the DNA triplet CAA is the code for an amino acid called valine, and CGA is the triplet for an amino acid called alanine. If these triplets form part of a gene they will cause a protein to form with the relevant amino acids in sequence, as outlined in diagram 2.8. Triplets are similar to the way in which Morse code uses a sequence of three dots or dashes to specify a letter.

A gene consists of a long stretch of triplets or codons that code for a specific sequence of amino acids, allowing for the production of a particular protein.

Replication of DNA

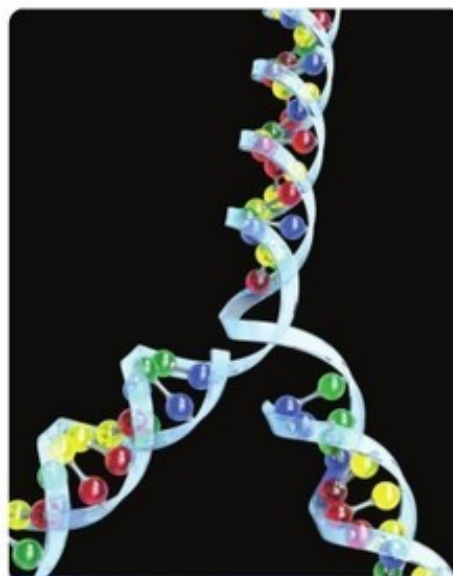
At the end of mitosis each new cell has single-stranded chromosomes. Before these cells can divide, the DNA in each chromosome must produce an exact copy of itself. This means that the single-stranded chromosomes must become double-stranded chromosomes. This process is called DNA replication and it takes place in the nucleus during interphase (see diagram 2.10).

The genetic code is the sequence of bases in DNA that provide the instruction for a cell (using RNA) to form a protein.



2.8 DNA triplet code for particular amino acids

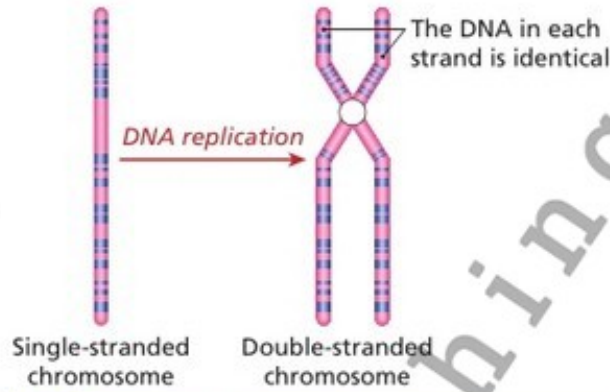
A triplet (or codon) is a sequence of three bases in DNA (or RNA) that act as a code for an amino acid.



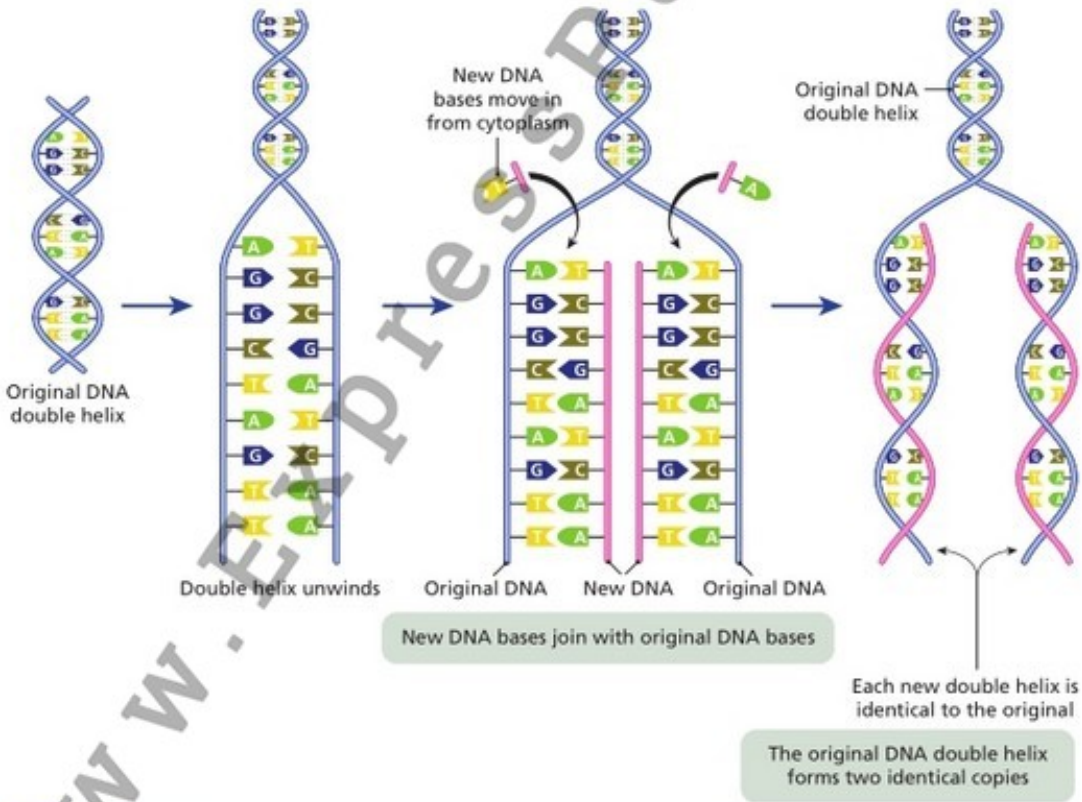
2.9 DNA replication

Mechanism of DNA replication (see diagram 2.11)

- 1 The double helix unwinds (or uncoils).
- 2 An enzyme breaks the bonds between the base pairs. The two strands of the original double helix separate.
- 3 DNA bases that are normally present in the cytoplasm enter the nucleus. The incoming bases attach to the exposed complementary bases (i.e. base pairing occurs).
- 4 In this way, each side of the DNA molecule acts as a mould or template for the new DNA that is formed.
- 5 Each new double strand rewinds to form a double helix. Note that each new DNA double helix is:
 - ▶ Half new DNA and half original DNA
 - ▶ Identical to the original DNA double helix and to the other new double helix formed.



2.10 An overview of DNA replication

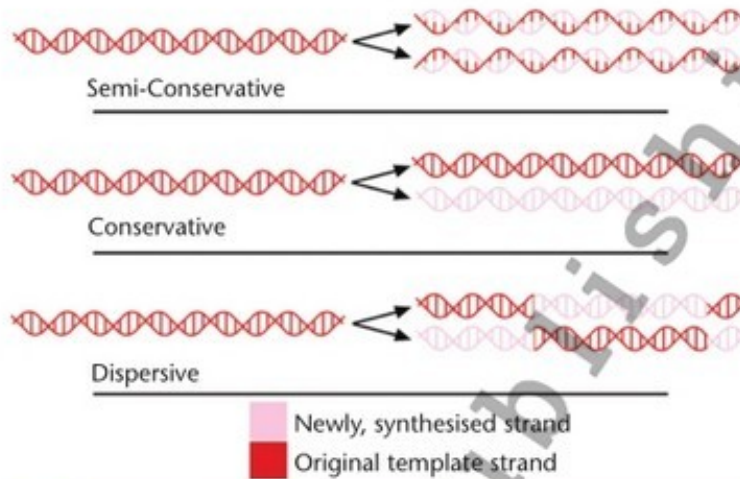


2.11 DNA replication

Discovery of how DNA replicates

When the structure of DNA was first discovered, there were three different theories of how it could replicate in cells. These were called conservative, semi-conservative and dispersive and are summarised in diagram 2.12.

Two scientists called Meselson and Stahl carried out an experiment to determine which of these three methods occurs in cells.



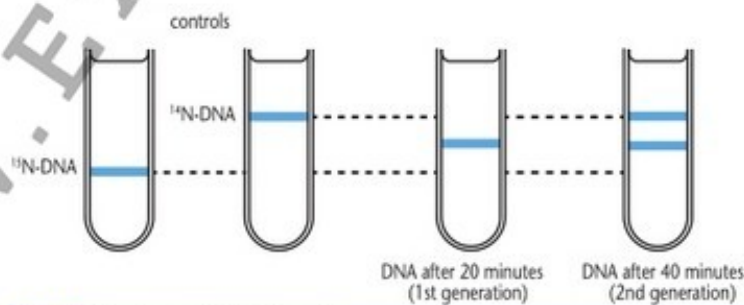
2.12 Three postulated methods of DNA Replication

Meselson and Stahl carried out their experiments on the DNA replication of *E.coli* bacteria. They grew bacteria in medium that contained ^{15}N , which is a heavier isotope of nitrogen than the form, ^{14}N , that occurs naturally. The bacteria take in this ^{15}N and incorporate it in their DNA, which becomes heavier and more dense as a result. The bacteria were left in the medium with ^{15}N until all of the ^{14}N in their DNA had been replaced with ^{15}N .

The DNA was extracted and its density measured.

The bacteria were then transferred to medium containing the more normal ^{14}N and allowed to grow. Each time they would make new DNA, the ^{15}N in their old DNA would be replaced with ^{14}N . Their DNA would become lighter and less dense as a result.

Meselson and Stahl knew that *E.coli* bacteria would replicate every 20 minutes. Each time the bacteria divided and formed a new generation, the DNA was extracted and its density measured. Diagram 2.13 shows Meselson and Stahl's results.



2.13 Meselson and Stahl's results

The results prove that replication is semi-conservative. All of the DNA after 1 generation had a density intermediate between that of ^{15}N -DNA and ^{14}N -DNA. This means that one old strand has ^{15}N and the other new strand has ^{14}N . If replication had been conservative, then there

would be two bands of DNA after one generation: one from the old DNA that had ^{15}N in both its strands and one from the new DNA that had ^{14}N in both its strands. Two bands would also be seen at 1 generation if replication was dispersive.

The second-generation DNA produced two bands in the experiment. One was in the same position as the intermediate band from the first generation and a second band in the position of the lighter ^{14}N molecule. This result indicated that DNA was being replicated semi-conservatively.

Chargaff's rule

In DNA, guanine can only bond to cytosine and adenine can only bond to thymine. This means that the quantity of cytosine and guanine must be equal to each other and the quantity of adenine and thymine must also be equal to each other. These quantities are usually expressed as percentages.

For example, a molecule of DNA is known to contain 22% adenine. From this, we can calculate the percentages of all the other bases in the DNA as follows:

If adenine = 22%, then by Chargaff's rule, thymine must also be 22%.

Adenine and thymine together make up 44% (22 + 22) of the DNA.

The remainder of the DNA, 56% (100 – 44) is made up from cytosine and guanine.

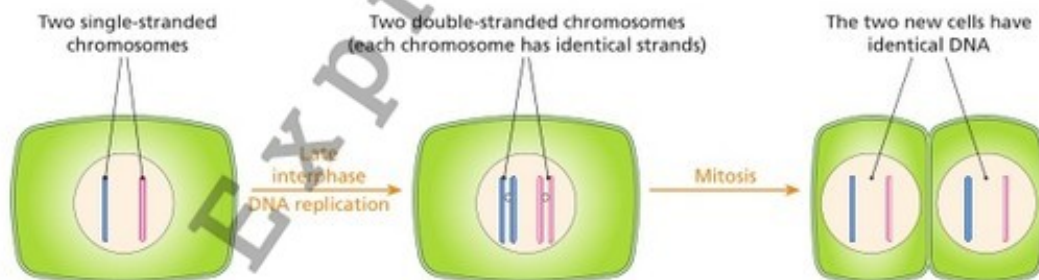
By Chargaff's rule, the percentages of cytosine and guanine must be equal, so they are both 28% (56 ÷ 2).

Significance of DNA replication

Each new DNA double helix (chromosome) will have exactly the same sequence of bases as the original (as shown in diagram 2.11).

DNA is able to produce **exact** copies of itself (hence the term 'replication' is used to describe its manner of reproducing). This allows the same DNA to be passed on to each new generation of cells, as outlined in diagram 2.14.

For example, a human zygote is a single cell with 46 chromosomes. These chromosomes contain a certain sequence of bases. The same sequence of bases is passed on, in the form of new chromosomes, to each body cell in a person due to DNA replication (and mitosis).



2.14 The significance of DNA replication

DNA profiling

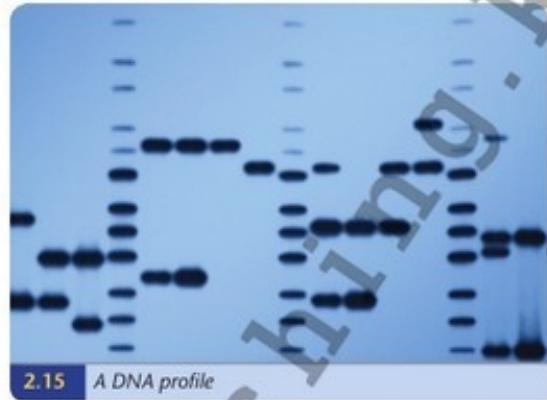
A DNA profile (also called a DNA or genetic fingerprint) is a method of making a unique pattern of bands from the DNA of a person, which can then be used to compare with the DNA profile of another person.

Method of preparing a DNA profile

Preparing a DNA profile involves four steps:

- 1 The DNA is released from cells
- 2 The DNA is cut into fragments of different lengths
- 3 The DNA fragments are separated according to their sizes
- 4 The patterns produced by the fragments are compared or analysed.

The following describes these four procedures in more detail.



2.15 A DNA profile

1. DNA is released

In order to produce a DNA profile, cells are broken down to release their DNA (as in Experiment 2.1).



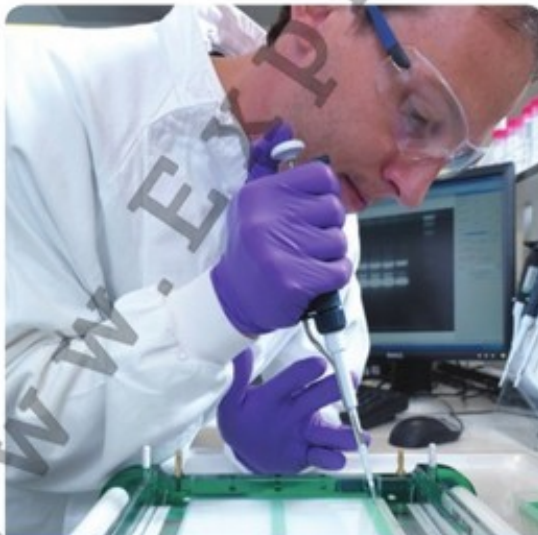
If the amount of DNA available is too small to work with, it can be increased or amplified. A common technique used to amplify small quantities of DNA is a process called the polymerase chain reaction (PCR).

2. DNA is cut into fragments

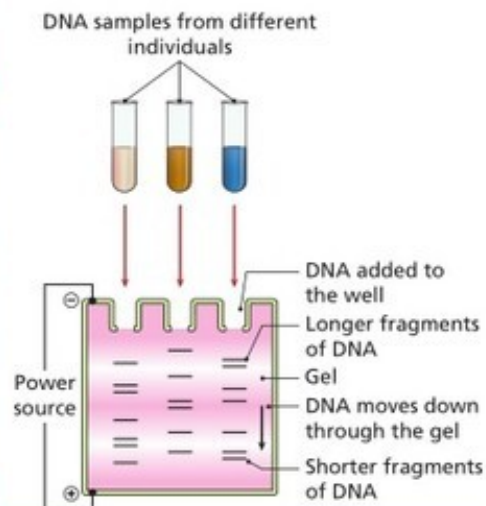
The isolated DNA is cut into fragments using special enzymes. These enzymes are called **restriction enzymes**. They were first isolated from bacteria, where they are used to destroy the DNA of invading viruses.

Different restriction enzymes cut DNA at specific base sequences. For example, one restriction enzyme will always cut DNA at the base sequence GAATTC, whereas another only cuts at the sequence GATC.

The sections of DNA that are cut will be of different lengths because the base sequences being cut may be close together or far apart on the DNA strands.



2.16 Adding DNA to a gel for electrophoresis



2.17 Separating DNA by gel electrophoresis

3. The fragments are separated

The sections of DNA that have been cut are separated on the basis of their size. They are separated by a process called gel electrophoresis. This involves placing the invisible DNA fragments in a small glass tank containing a sugar-based gel. An electric current is applied along the gel. The current draws the negatively charged DNA to one end of the gel.

Small DNA fragments move faster through the porous gel than do the larger fragments. In this way bands of small fragments are separated from bands of larger fragments.

When the electrophoresis is finished a permanent record of the results is obtained. This may involve adding radioactive material, which combines with DNA fragments to produce a fluorescent image. A photographic copy of the final pattern of DNA bands is then obtained.

4. Patterns are compared

In the same way that no two people have the same fingerprints, it is highly unlikely that any two people will have the same DNA profile (unless they are identical twins).

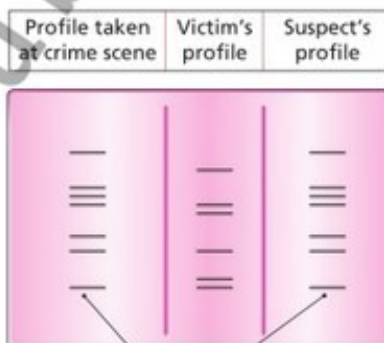
If the pattern of bands from two different DNA samples is the same, then the two samples must have come from the same person.

Applications of DNA profiles (DNA fingerprints)

Crime

DNA profiles are often used in forensic (legal) cases. If biological material such as blood, hair, saliva or semen is left at the scene of a crime it can be collected and a DNA profile prepared.

- If the pattern of the DNA profile from the crime scene is compared with those of the victim and a suspect, it may be seen that it matches that of the suspect but not that of the victim. This would be strong evidence to associate the suspect with the crime scene.
- Of course the profile may not match that of the suspect, which might eliminate the suspect from the inquiry.



These profiles match, indicating the suspect was at the crime scene

2.18 Forensic use of DNA profiles

Forensic medicine is the way in which medical knowledge is used in legal situations.

Medical

DNA profiles can be used to determine whether a particular person is, or is not, the parent of a child. In this way the paternity (father) or maternity (mother) can be established. This information can apply in property or financial inheritance cases, or in immigration cases where a person can enter a country if his/her parent or child is already in that country.

To decide if a man is the father of a child, blood samples are taken from the child, the mother and the man. DNA profiles are prepared and examined.

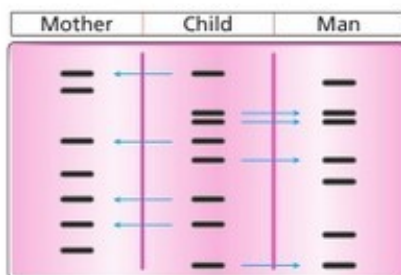


The bands differ in thickness due to there being more or less bands of this length present, e.g. a thick band represents many DNA fragments of a particular length. This is visible in diagram 2.19.



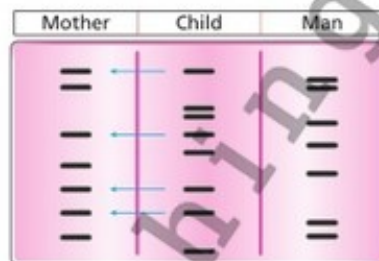
2.19 DNA profiles for a family: M represents the mother, C the children and F the father

- If all the bands in the child's profile match with bands in either the mother's or the man's profile, then the man is shown to be the natural father of the child (see diagram 2.20).
- If some of the child's bands match those of the mother, but the rest do not match with the man's bands, then the man is not the father of the child (see diagram 2.21).



All the child's bands match with the mother's or the man's.
The man is the father of the child.

2.20 Using DNA profiles to determine the father of a child: a match



Some of the child's bands match with the mother's. The rest of the child's bands do not match with the man's.
The man is not the father of the child.

2.21 Using DNA profiles to determine the father of a child: no match

Genetic screening

Sometimes the process of DNA replication does not work exactly as it should. In these cases, a gene (or a number of genes) may be incorrectly copied. In addition, DNA can be altered by mutations.

If genes are altered in any way they will not carry the correct code for the protein that they were intended to produce. This may have severe effects on a person who inherits such genes.

Genetic screening often involves adding a radioactive section of DNA (called a DNA, or genetic, probe) to a sample of DNA from the person being tested. The DNA probe will only attach to a normal gene. If the probe does not attach then the gene is altered.

Genetic screening can be carried out in two main ways: adult screening and foetal screening.

Adult screening

Screening is sometimes carried out on adults who, although they do not suffer from a genetic disorder, may carry a defective gene in each of their cells. People who carry defective genes, without having the disorder themselves, are said to be carriers for the condition.

It is now possible to identify (or genetically screen) individuals who are carriers for many disorders such as sickle cell anaemia, the most common form of cystic fibrosis, and the likelihood of getting heart disease or some forms of cancer.

The benefits of these tests is that it gives people information regarding the chances of them having a child with the disorder or it allows them to prepare for a disease that might affect them.

Genetic disorders caused by defective genes include:

- Albinism (in which the pigment melanin cannot be made)
- Cystic fibrosis (in which there is a build-up of mucus in the lungs and intestines)
- Haemochromatosis (in which too much iron accumulates in the body and has to be removed by regular bleeding)
- Cancer.

Genetic screening means testing DNA for the presence or absence of a particular gene or an altered gene.

Embryonic or foetal screening

In this type of screening, cells can be removed from the embryo, placenta or the fluid around the foetus. These cells can be tested to detect if the embryo or foetus has any one of a number of genetic disorders.



2.22 Obtaining fluid containing foetal cells for genetic screening



Ethics of genetic screening

Ethics relates to whether behaviour is proper or improper. Genetic screening may cause ethical problems.

If the results of genetic tests become public, the people concerned may suffer embarrassment or be treated unfairly. For example, they may be isolated and treated as if they had a disease even if they do not. Employers and insurance companies may be reluctant to get involved with them.

Another issue is whether a person would wish to know that they have a genetic disorder that will develop in later years. This is especially problematic if the disorder is untreatable.

In relation to genetic screening in the embryo or foetus, would knowledge of a genetic disorder help to prepare the family for the future, or encourage a termination of the pregnancy?

RNA (ribonucleic acid)

DNA and RNA are both nucleic acids. However, RNA differs from DNA in the following ways:

- RNA is based on the sugar ribose (whereas DNA is based on the sugar deoxyribose)
- RNA (ribonucleic acid) consists of four bases, like DNA. However, RNA contains the base uracil instead of thymine. This means that the bases in RNA are A, U, G and C. The bases A and U are complementary, as are the bases G and C.

The difference between the bases in DNA and RNA are simplified in diagram 2.24.

- The sequence of bases in RNA is determined by the sequence of bases in DNA. The bases in RNA are complementary to those in a section of DNA. For example, if a strand of DNA has the sequence GGAATC, then the RNA produced will have the sequence CCUUG.
- RNA is a single-stranded molecule, unlike DNA, which is a double strand (helix).
- RNA can move out of the nucleus into the cytoplasm. DNA is always in the nucleus.



2.23 Base pairs in RNA



2.24 Difference between bases in DNA and RNA

Structural differences between DNA and RNA

DNA	RNA
1 Contains the sugar deoxyribose	Contains ribose
2 The bases are AT GC (thymine)	The bases are AU GC (uracil)
3 Double-stranded (i.e. double helix)	Single-stranded

Types of RNA

There are several different types of RNA used in cells. Three of the most important are used in making proteins in cells.

Messenger RNA, or mRNA, is used to carry the message for protein synthesis from the nucleus out into the cytoplasm. It is single stranded without any defined structure as it is only the sequence of bases that is important.

Transfer RNA, or tRNA, is used to bring specific amino acids for making the new protein. It is also single stranded but has a well defined structure unlike mRNA. This structure is sometimes called “clover-leaf” and can be seen in diagram 2.25. The structure is held by parts of the tRNA being folded back on themselves and hydrogen bonded by complementary base pairing.

Ribosomal RNA, or rRNA, is part of the structure of ribosomes, as the name suggests. Ribosomes are approximately 60% rRNA and 40% protein by mass. The genes that code for rRNA are one of the only types of genes that are found in all cells!

RNA has some other functions in cells and can also be found in some viruses instead of DNA.



2.25 Structure of Transfer DNA or tRNA

Detailed structure of DNA

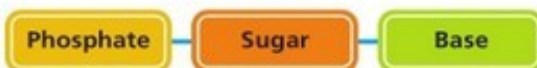
The structure of DNA was worked out by James Watson and Francis Crick in 1953. They shared the Nobel prize in 1962 with Maurice Wilkins for their discovery. Their findings were based on the earlier research of Rosalind Franklin, who unfortunately died in 1958.

The discovery of the structure of DNA is considered to be one of the outstanding advances in biology in the 20th century. DNA is made up of units called nucleotides. These are arranged into very long chains called polynucleotides.

The detailed structure of DNA can be considered under three headings: nucleotides, base pairs and double helix.

Nucleotides

A nucleotide consists of three parts: a phosphate group, a sugar and a nitrogen-containing base. These are linked together as shown in diagram 2.26.

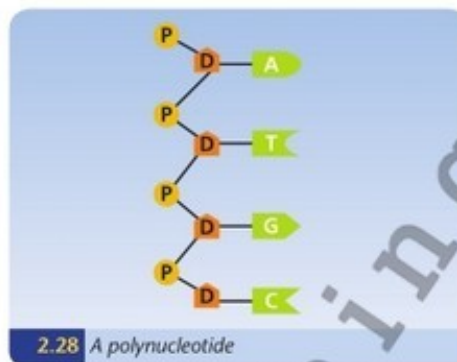
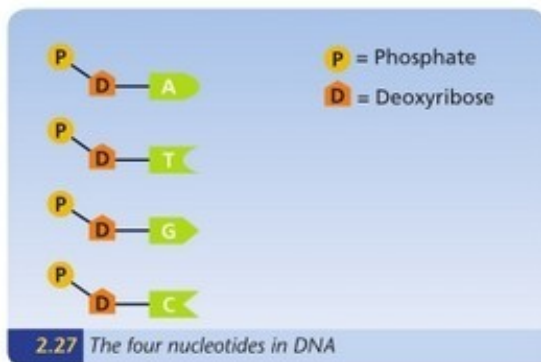


2.26 Structure of a nucleotide

The sugar in DNA is deoxyribose (i.e. a five-carbon sugar, similar to ribose but lacking an oxygen atom). RNA contains the sugar ribose.

The phosphate group is PO_4 , but this is normally represented as P. The phosphate and deoxyribose groups form the sides of the DNA strand.

As there are four bases this means that there are four distinct nucleotides. The nucleotides join together, with a bond between the phosphate group of one and the sugar of the next, forming a polynucleotide.



Purines and pyrimidines

Of the four nitrogenous bases in DNA, two are classified as purines (double-ring molecules) and two as pyrimidines (single-ring molecules).

The purine bases can be remembered by the phrase: The Attorney General is Pure

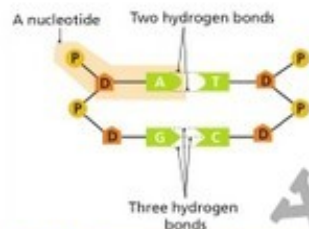
The purines are adenine (A) and guanine (G).
The pyrimidines are thymine (T) and cytosine (C).

Base pairs

The bases join together in a very specific manner. Adenine and thymine each form two weak hydrogen bonds. This allows them to bond together. In a similar way, guanine and cytosine each form three hydrogen bonds, so they can pair together.

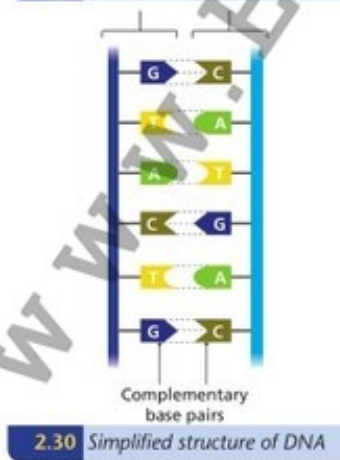
The pyrimidines can be remembered because: the pyrimidines contain the letter 'y' i.e. thymine and cytosine.

The bases can be thought of as having opposite or complementary shapes, just like adjoining pieces of a jigsaw. Note that each base pair has a purine and a pyrimidine.



The forces holding the bases together are **hydrogen bonds**. These are weak bonds formed when a slightly positive hydrogen is attracted by another slightly negative atom (e.g. nitrogen or oxygen).

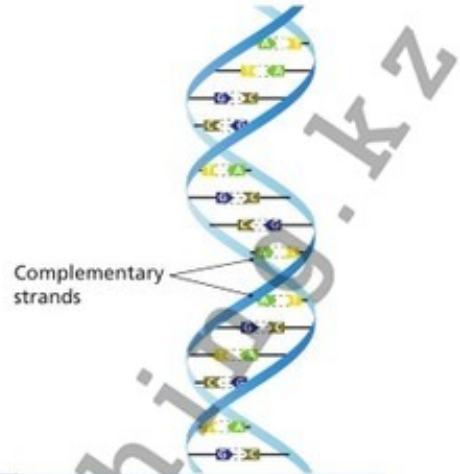
Very often the phosphates and sugars are left out when drawing a simplified version of DNA. In this case, only the base pairs are shown, as seen in diagram 2.30.



2.31 Watson and Crick with their model of DNA in 1953

Double helix

Francis Crick and James Watson discovered that DNA consisted of two helical or spiral chains of polynucleotides, as shown in diagram 2.32. The outside strands of the double helix are made of deoxyribose and phosphate. The 'rungs' of the molecule are the base pairs on the inside.



2.32 Double helix of DNA

Protein synthesis

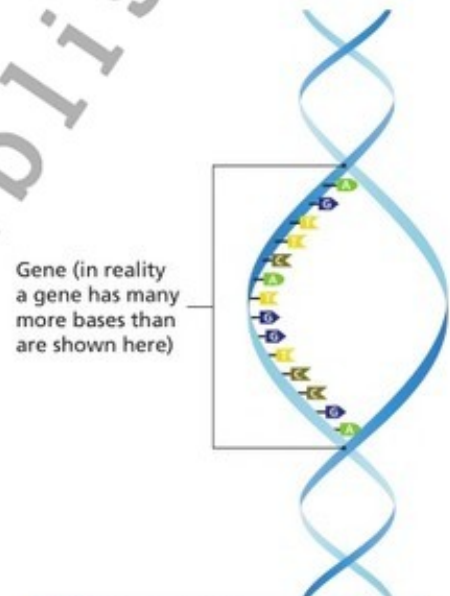
The process of protein formation proceeds as follows.

Initiation: starting the process

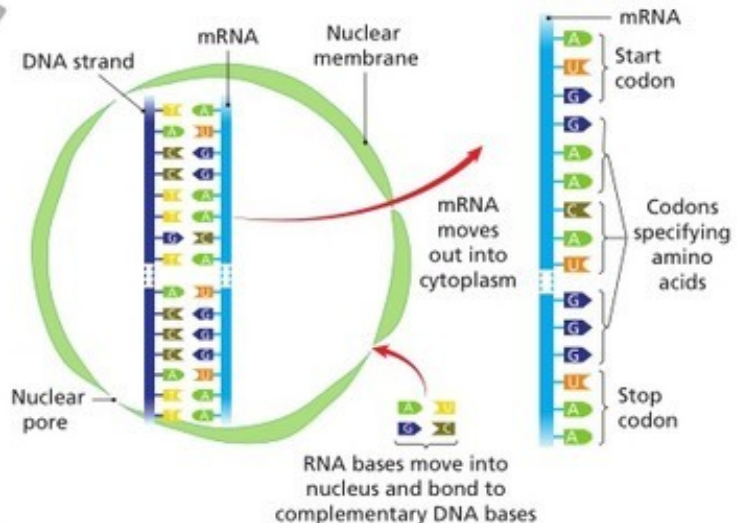
- 1 The DNA double helix unwinds at the site of the gene that is going to produce a protein.

Transcription: rewriting the code from DNA to RNA

- 2 RNA bases, which are present in the cytoplasm, move across the nuclear membrane. The RNA bases will match up with the complementary bases on the DNA strand.
- 3 The enzyme RNA polymerase causes the sequence of RNA bases to join together to form messenger RNA (mRNA). Each mRNA molecule has complementary bases to those on the DNA strand from which it was transcribed. A sequence of three bases of DNA or RNA is called a triplet or codon. Codons may cause three possible outcomes.
 - ▶ A start codon indicates the beginning of a gene (but is not involved in the production of the protein)
 - ▶ Most codons in a gene specify particular amino acids
 - ▶ A stop codon indicates the end of a gene (but is not involved in the production of the protein).
- 4 Every gene has **one** start codon, **many** codons specifying amino acids and **one** stop codon.



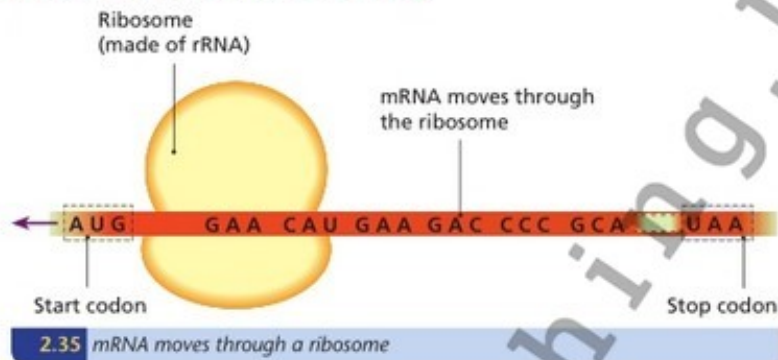
2.33 The double helix unwinds at the site of the gene



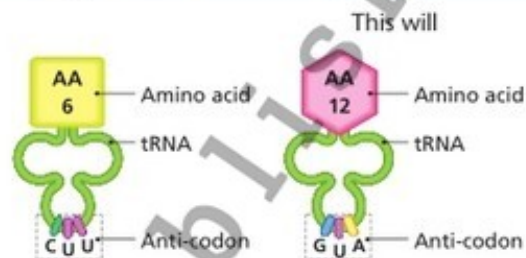
2.34 Transcription

Translation: the production of a protein according to the RNA code

- 5 mRNA moves from the nucleus to the cytoplasm.
- 6 Ribosomes are made up of ribosomal RNA (rRNA) and protein.
- 7 The mRNA strand forms weak bonds with the rRNA in a ribosome.

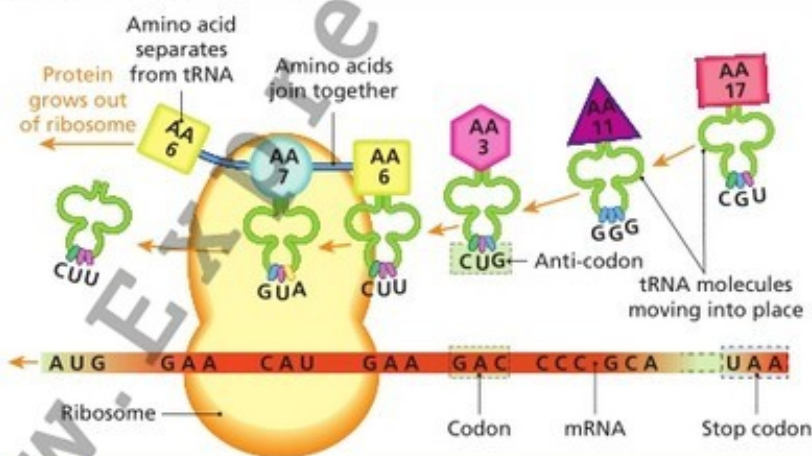


- 8 The cytoplasm contains a supply of transfer RNA (tRNA) molecules. Each tRNA carries:
 - ▶ A special triplet or anticodon
 - ▶ A particular amino acid, which is specific to the anticodon.



- 9 tRNA molecules are attracted to the mRNA that is in the ribosome. Each anticodon on a tRNA is complementary to a codon on the mRNA. The tRNA molecules enter the ribosome.
- 10 The first tRNA molecule will be attracted to the mRNA just after the start codon (see left-hand side of diagram 2.37). In doing this it brings a particular amino acid to the ribosome.

An anticodon is a sequence of three bases (a triplet) on tRNA that are complementary to a sequence of three bases on mRNA.



- 11 In the ribosome, amino acids are detached from the tRNA molecule and are bonded together to form part of the new protein.
- 12 tRNA molecules leave the ribosome without any attached amino acids. As they leave they pull the mRNA strand through the ribosome.
- 13 tRNA molecules continue to bind with mRNA until a stop codon is reached. At this point:
 - ▶ The mRNA code sequence is complete
 - ▶ The new protein is produced.

- 14 Once the protein is formed it folds to allow it to have the correct shape.

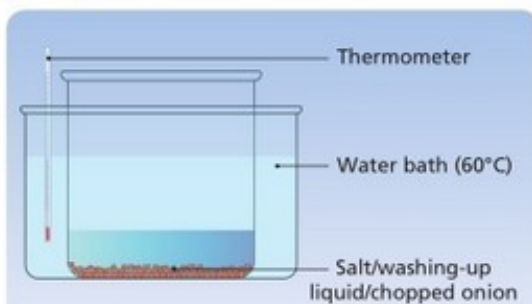
Functions of the three types of RNA	
Type of RNA	Function
mRNA (m = messenger)	<ul style="list-style-type: none"> Complementary strand to DNA Carries instruction for the production of a protein from DNA to a ribosome
tRNA (t = transfer)	<ul style="list-style-type: none"> Has a complementary anticodon to mRNA codon Carries an amino acid to the ribosome
rRNA (r = ribosomal)	<ul style="list-style-type: none"> Forms part of the structure of a ribosome Forms a weak bond with mRNA in the ribosome.

Experiment 2.1 To isolate DNA from a plant tissue

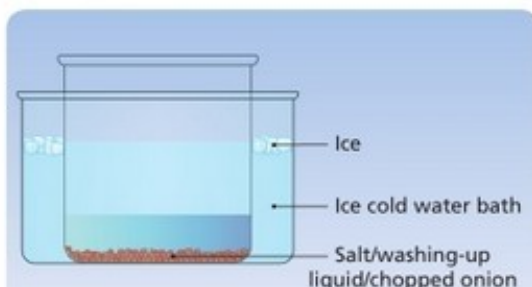
- Add some sodium chloride (salt) to a small volume of washing-up liquid dissolved in water (the salt will cause the DNA molecules to clump together, and the washing-up liquid will dissolve the cell and nuclear membranes and release DNA from the cells).
- Cut an onion (or kiwi fruit) into small cubes (this allows the washing-up liquid to reach more cells).
- Add the chopped onion to a beaker containing the salt/detergent solution and stir the mixture.
- Put the beaker in a water bath at 60°C for 15 minutes. (This temperature inactivates (denatures) enzymes that would normally digest DNA. If left any longer than 15 minutes DNA itself would break down.)
- Cool the mixture by placing the beaker in an ice water bath for 5 minutes, stirring frequently (this slows down the breakdown of the DNA).
- Pour the mixture into a domestic food blender and blend it for only 3 seconds on high speed. (This breaks down the cell walls and releases DNA. Blending it for too long would break down the DNA strands.)



2.38 Steps 1 to 3

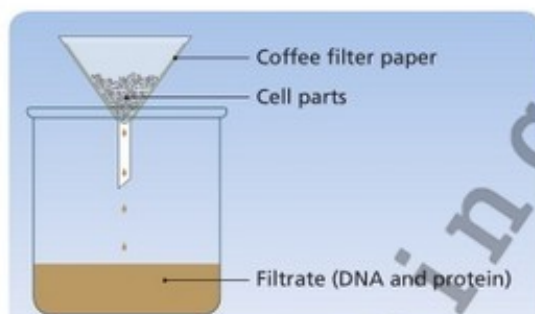


2.39 Step 4

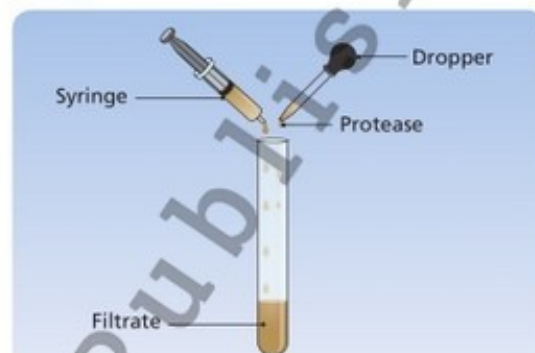


2.40 Step 5

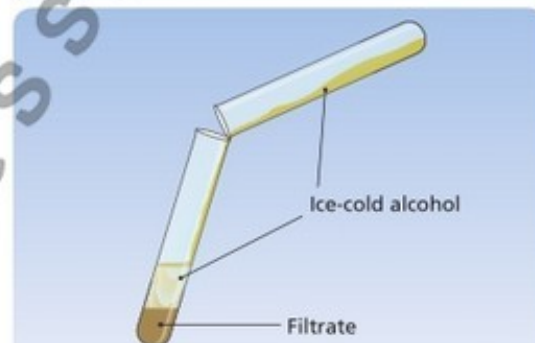
- 7 Filter the mixture through coffee filter paper into a second beaker. Do not add the foam from the top of the mixture to the filter paper. (Cell parts are retained in the filter paper. The filtered material, called filtrate, contains DNA and proteins. Normal laboratory filter paper is not used as its pores are too small and the process would be very slow.)
- 8 Use a syringe, without a needle, to place some of the onion filtrate into a boiling tube.
- 9 Add a few drops of protease enzyme (such as pepsin) to the contents of the boiling tube and mix well (the protease breaks down the proteins around the DNA).
- 10 Pour some ice-cold ethanol or methylated spirits (stored in a freezer overnight) carefully down the side of the boiling tube. The ethanol should form a layer on top of the onion filtrate. (Alcohol removes water from DNA, which causes DNA to float to the top of the water. DNA is insoluble in ice-cold alcohol and so it precipitates at the alcohol–filtrate boundary. The DNA forms white threads at the alcohol–filtrate junction).
- 11 Gently twist a small glass rod or a wire loop in the alcohol. Strands of DNA should attach to the rod or wire. Do not mix the two layers or damage the DNA, which is very easily broken (DNA forms a clear mesh of what looks like stringy mucus, as shown in diagram 2.4).



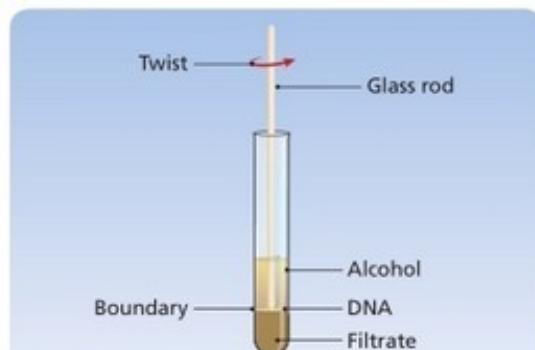
2.41 Step 7



2.42 Steps 8 and 9



2.43 Step 10



2.44 Step 11

Questions on Module 2

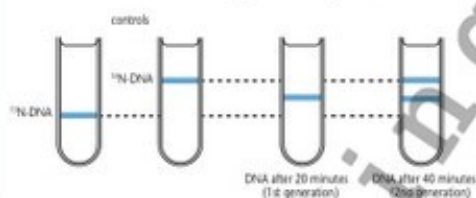
- 1 (a) What is heredity?
(b) Name the key chemical that is inherited by organisms.
(c) Name three inherited characteristics in (i) Humans
(ii) Plants.
- 2 (a) Distinguish between a gene and gene expression.
(b) What do you understand by 'characteristics'?
(c) Characteristics are formed by the interaction of two factors. Name these factors.
- 3 (a) Name the materials of which chromosomes are made.
(b) Give a function for each of these materials.
(c) Show, by means of a diagram, the relationship between genes and chromosomes.
(d) Explain what is meant by (i) coding, and (ii) non-coding, DNA.
- 4 (a) What do the letters 'DNA' stand for?
(b) What name is given to the overall shape of DNA?
(c) Explain how a very long DNA molecule can fit into a tiny nucleus.
- 5 One strand of a DNA molecule has the base sequence GATTCGTA.
(a) What is the sequence of bases on the complementary DNA strand?
(b) How many triplets are in this sequence of DNA?
- 6 (a) Name the bases in DNA.
(b) What is a DNA triplet?
(c) How do triplets relate to genes?
- 7 State two places in a chromosome where non-coding DNA may occur.
- 8 (a) What is meant by DNA replication?
(b) What is the importance of DNA replication?
(c) At what stage of the cell cycle does DNA replication occur?
(d) Where in a cell does DNA replication occur?
- 9 What is genetic screening?
- 10 Outline two structural differences between DNA and RNA.
- 11 If a stretch of DNA has the base sequence ATTGGCATT, what will the base sequence be on the complementary RNA strand?
- 12 What material do genes produce in order to control the activities of a cell?
- 13 (a) What is meant by base pairing in DNA?
(b) Draw a diagram of a stretch of DNA comprising eight nucleotides to indicate base pairing involving all the possible base pairs (**there is no need to draw a double helix**).
(c) Who discovered the double helix structure of DNA?
- 14 (a) What is the full name for RNA?
(b) Name three types of RNA.
(c) State two places in a cell where RNA might be found.
(d) What is meant by the phrase 'DNA codes for messenger RNA'?
- 15 In isolating DNA from plant tissue, give a reason for each of the following:
(a) Using washing-up liquid
(b) Using salt
(c) Heating the salt, detergent and chopped tissue to 60°C
(d) Using ice-cold water
(e) Not blending the tissues for too long
(f) Using protease enzyme
(g) Adding ice-cold alcohol.
- 16 Choose which of the options (i), (ii), (iii) or (iv) represents the correct answer in each case below.
(a) DNA is insoluble in:
(i) Washing-up liquid
(ii) Boiling water
(iii) Salt water
(iv) Ice-cold ethanol.
(b) DNA fragments can be obtained using:
(i) Bacteria
(ii) Protein synthesis
(iii) Restriction enzymes
(iv) Ribosomes.
(c) DNA fingerprinting can be used to determine if a person:
(i) Has an infectious disease
(ii) Is or is not the parent of a child
(iii) Has faulty RNA
(iv) Is a carrier of a genetic disease.
- 17 Three nucleotides make up one triplet of bases in DNA. There are four different bases. Work out how many possible triplets can be made from three bases.

- 18 Under what circumstance could two people have **exactly** the same genetic profile?
- 19 All the human chromosomes in one cell have a total of 3 billion (3×10^9 or 3,000,000,000) base pairs of DNA. Cells use a lot of cellular energy to replicate all of this DNA when the cells need to form new copies of themselves. More than 95% of the 3 billion base pairs is non-coding DNA. Suggest why the term 'junk DNA' sometimes used for non-coding DNA is not appropriate.
- 20 Draw how the bands of DNA would have appeared in the first generation of Meselson and Stahl's experiment if DNA replication had been:

(a) Conservative

(b) Dispersive

Refer to this diagram for your answer.



2.45 Meselson and Stahl's results

- 21 A double stranded DNA molecule is found to be composed of 24% thymine. Calculate the percentages of the other three bases in this DNA molecule.

Module 3 Cell structure

Learning objectives

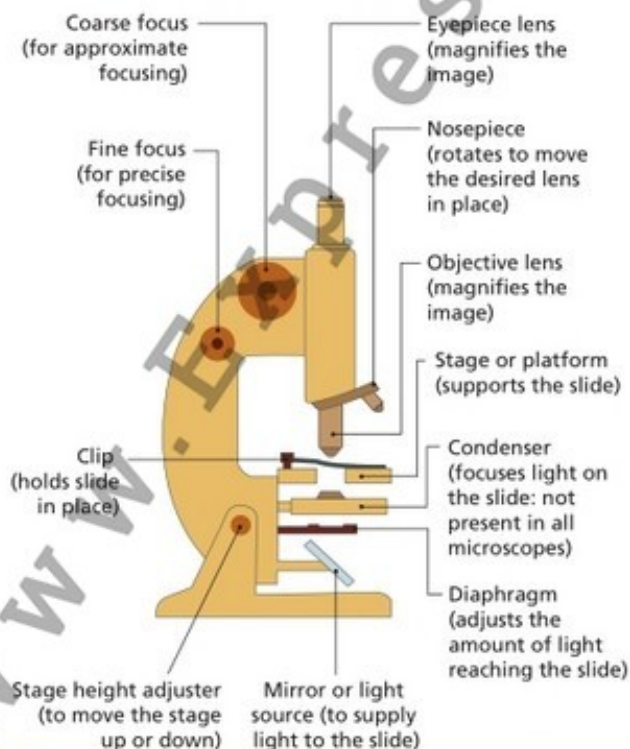
- Explain the features of structure and functions of the cell organelles visible under the electron microscope (10.4.2.1)
- Establish a connection among the structure, properties and functions of the cell membrane, using the fluid mosaic model (10.4.2.2)
- Compare features of the structure and function of prokaryote and eukaryote (10.4.2.3)

Microscopes

A **compound microscope** uses two lenses, an objective lens and an eyepiece lens, as shown in diagram 3.1. The total magnification of the image is calculated by multiplying the power of the two lenses (see table below).

Magnification		
Eyeiece lens	Objective lens	Total
x5	x10	x50
x10	x40	x400

Compound microscopes use light to show the image. The maximum magnification that can clearly be achieved is about $\times 1000$ for a compound microscope.

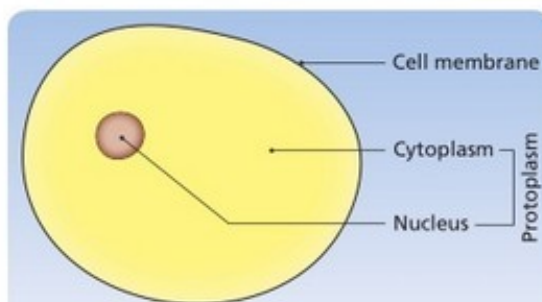


3.1 Parts of a compound light microscope and their functions



3.2 A light microscope

Cell structure as seen using a light microscope



3.3 Animal cell as seen under a light microscope

Animal cells

Animal cells are surrounded by an outer membrane, called the cell or plasma membrane. This membrane surrounds the protoplasm.

The protoplasm of a cell is made up of the nucleus and the surrounding cytoplasm.

Many of the reactions in a cell take place in the cytoplasm.

The protoplasm is all the living parts of a cell.

The cytoplasm is the living material in a cell outside the nucleus.

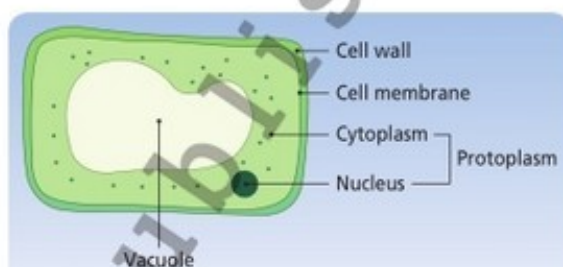
Plant cells

Plant cells are enclosed by a rigid **cell wall** made of cellulose. Cellulose is a strong structural carbohydrate (or polysaccharide) and is the main component in paper and cotton wool.

The cell wall gives the cell strength and makes it less flexible. The **cell membrane** is usually found just inside the cell wall. The wall and membrane are often so close that the membrane is not seen clearly.

Vacuoles contain a fluid called cell sap. This is a solution of salts, sugars and pigments.

Plant cells also have a **nucleus** and **cytoplasm**. Plant cells that are green contain structures called **chloroplasts**. These are the structures in which photosynthesis takes place.



3.4 Plant cell as seen under a light microscope

The vacuole helps to give the cell strength and shape and may also store materials.

Cell ultrastructure

Electron microscopes use a beam of electrons instead of light. As electrons are invisible, the image is often converted to an image on a TV screen.

There are two main types of electron microscope.

- A **transmission electron microscope** (TEM) sends a beam of electrons through a thin section of the specimen. This shows the internal structure of the specimen in great detail.
- A **scanning electron microscope** (SEM) uses a beam of electrons to provide a surface view of the specimen.

Ultrastructure is the detail of a structure as seen using an electron microscope.



3.5 An electron microscope

Electron microscopes can give magnifications of 250 000 and higher. In addition, they can produce very clear images. This makes electron microscopes ideal for observing very small structures.

Ultrastructure of a generalised cell

Fluid mosaic model of cell membranes

All membranes in biology are thought to have the same structure. They are composed of phospholipids and proteins.

The phospholipids, which have a water-loving phosphate group and a water-hating lipid group, are arranged into double layers (bilayers). The phosphates are on the exposed outer surfaces with the lipids in the middle.

In a membrane protein, molecules are completely or partially embedded in the phospholipid bilayer. Some of these proteins are attached to the bilayer; others are detachable and can move throughout the bilayer.

Functions of membranes

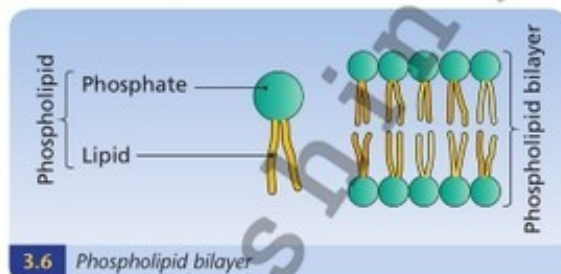
The functions or roles of membranes include to:

- Retain the cell contents
- Recognise molecules that touch them
- Control what enters and leaves the cell
 - ▶ Membranes can allow the free passage of some molecules and prevent the passage of others; in this way they are said to be selectively (or semi-) permeable. For example, water and oxygen can pass freely across a membrane, but sodium ions and large proteins have to be moved across using energy.
- Give some support to the cell.

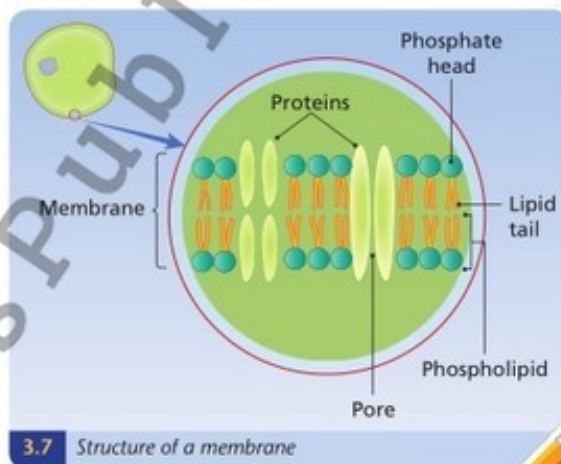
Nucleus

A nucleus is surrounded by a double membrane with numerous nuclear pores. These allow the controlled entry and exit of molecules into and out of the nucleus.

The nucleus contains strands of deoxyribonucleic acid (DNA). DNA is arranged into structures called chromosomes (*chroma* means 'colour', *soma* means 'body'; so-called because DNA readily absorbs many stains and becomes darkly coloured under the microscope).



3.6 Phospholipid bilayer



3.7 Structure of a membrane

The nucleus is the control centre of the cell.



3.8 A nuclear membrane showing nuclear pores (TEM)

Every organism has a definite number of chromosomes in each nucleus (e.g. humans have 46). Genes are located randomly along chromosomes.



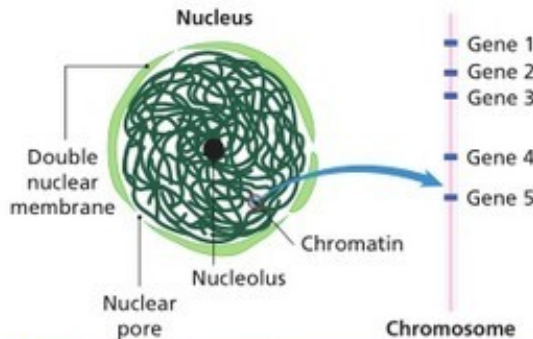
Humans are thought to have between 20 000 and 25 000 genes in each cell.

Genes are the structures that inform the cell how to make certain proteins. Genes control features such as the number of fingers, colour of eyes, production of enzymes and thousands more tasks. They are the units of inheritance.



Chromatin is the name given to chromosomes when they are elongated and not dividing.

When a cell is not dividing (i.e. most of the time), chromosomes are very elongated and interwoven. In this form they are called chromatin.



3.9 Nucleus and chromosome

Nuclear pores

Nuclear pores allow a type of RNA (ribonucleic acid) called mRNA (messenger RNA) to pass in and out of the nucleus. RNA is dealt with in Module 2.

Nucleolus

The nucleolus (plural nucleoli) is an area in the nucleus that stains very darkly.

The nucleolus makes ribosomes.

Cytoplasm

The cytoplasm is the jelly-like liquid in a cell that surrounds the nucleus.

A number of small bodies called **organelles** (such as mitochondria, chloroplasts and ribosomes) are suspended in the cytoplasm.

Mitochondria

Mitochondria (singular is mitochondrion) are the sites of respiration.

- Cells with many mitochondria (e.g. muscle and liver in animals, meristems in plants) produce lots of energy.
- Cells with few mitochondria (e.g. fat in humans, ground tissue in plants) produce less energy.

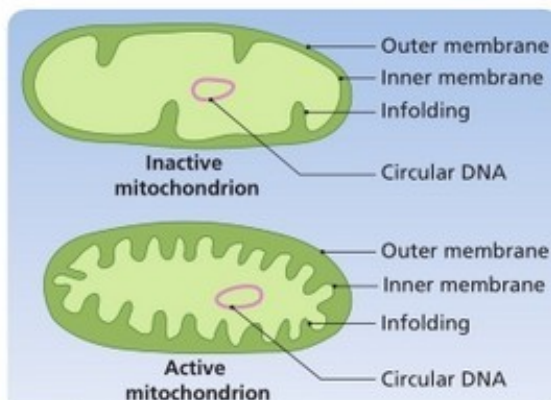
Mitochondria supply energy to the cell.

Mitochondria are surrounded by a double membrane. It is on the inner membrane, especially the infoldings, that energy is released. The more infoldings that are present the greater the surface area for cellular respiration, which results in the production of greater quantities of energy.

Each mitochondrion has its own loop of DNA referred to as circular DNA.



Active mitochondria convert to the inactive form if the cell rests for too long. This is why prolonged bed rest can cause tiredness when we try to resume normal life. Exercise causes the number of infoldings to increase again.



3.10 Ultrastructure of mitochondria



Chloroplasts (plants only)

Chloroplasts are surrounded by double membranes. They have membrane stacks, which contain the green pigment chlorophyll. They also have a loop of DNA.

Chloroplasts are green structures in plants in which photosynthesis takes place.



3.11 Chloroplasts (TEM): the dark circles are starch grains

Cell wall (plants only)

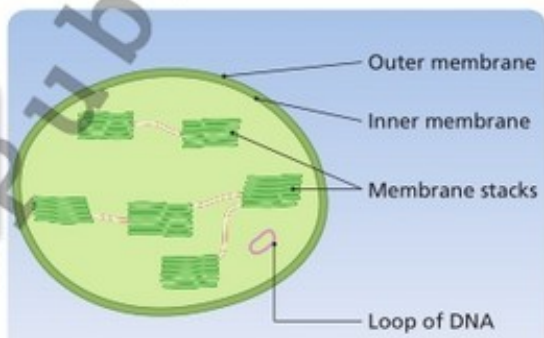
Plant cell walls are made of cellulose (which is a structural polysaccharide). Cell walls are fully permeable. This means that all molecules (big or small) can pass in or out through cell walls.

The function of cell walls is to support and strengthen the cell.

Ribosomes

Ribosomes are very tiny, bead-like structures. They are made of RNA and protein. They work by combining a sequence of amino acids to form a protein.

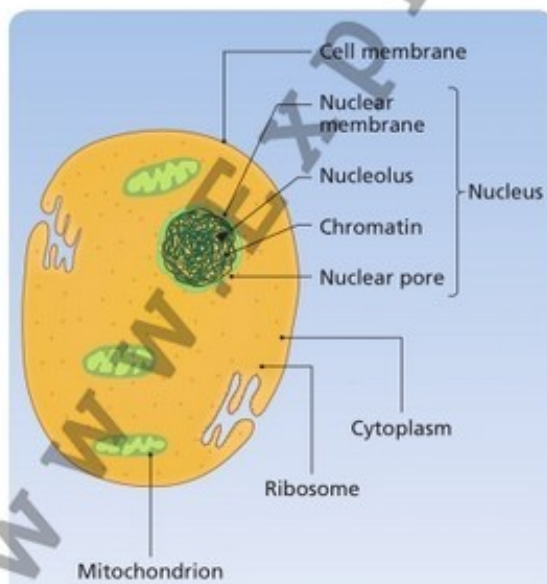
The function of ribosomes is to make proteins.



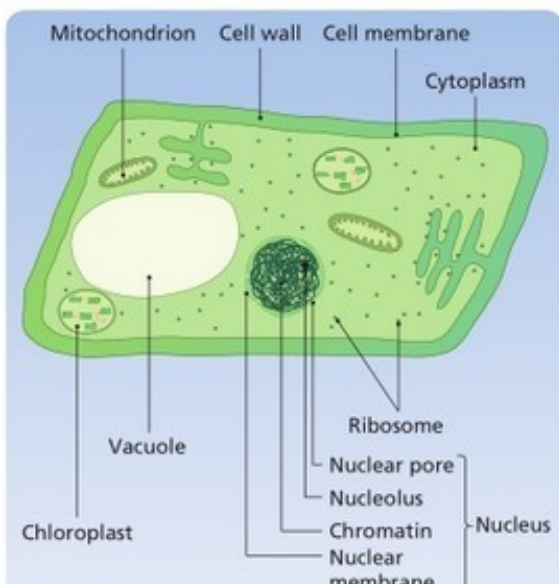
3.12 Ultrastructure of a chloroplast

Generalised cells

The ultrastructure of a general animal and plant cell is shown in diagrams 3.13 and 3.14.



3.13 Ultrastructure of an animal cell



3.14 Ultrastructure of a plant cell

Differences between plant and animal cells	
Plant cells	Animal cells
Have a cell wall	Do not have a cell wall
May have chloroplasts (containing chlorophyll)	Do not have chloroplasts (or chlorophyll)
Have a large vacuole	Do not have a large vacuole

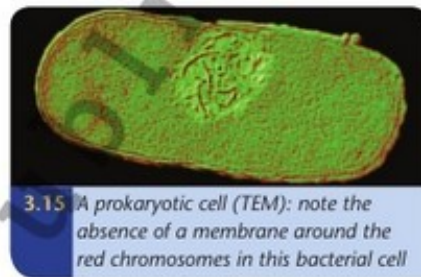
Prokaryotic and eukaryotic cells

Living things (also called organisms) can be placed into two categories depending on the structure and complexity of their cells: prokaryotes and eukaryotes.

Prokaryotic cells do not have a nucleus or membrane-enclosed organelles.

Prokaryotic organisms:

- Are single-celled
- Have a circular loop of DNA (not surrounded by a membrane, i.e. do not have a nucleus)
- Have small cells
- Do not have a membrane and enclosed structures such as mitochondria and chloroplasts
- Include bacteria.

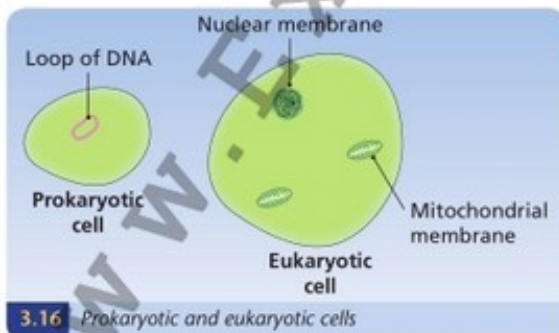


3.15 A prokaryotic cell (TEM): note the absence of a membrane around the red chromosomes in this bacterial cell

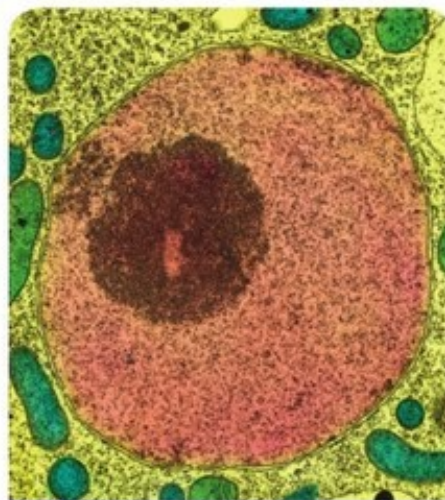
Eukaryotic cells have a nucleus and cell organelles, all of which are enclosed by membranes.

Eukaryotic organisms:

- Have a nucleus (i.e. DNA enclosed by a membrane)
- May have membrane-enclosed organelles such as mitochondria and chloroplasts
- Have large cells
- Include animals, plants and fungi
- Are more advanced than prokaryotes (i.e. life originated with prokaryotic cells and has evolved into eukaryotic cells).



3.16 Prokaryotic and eukaryotic cells

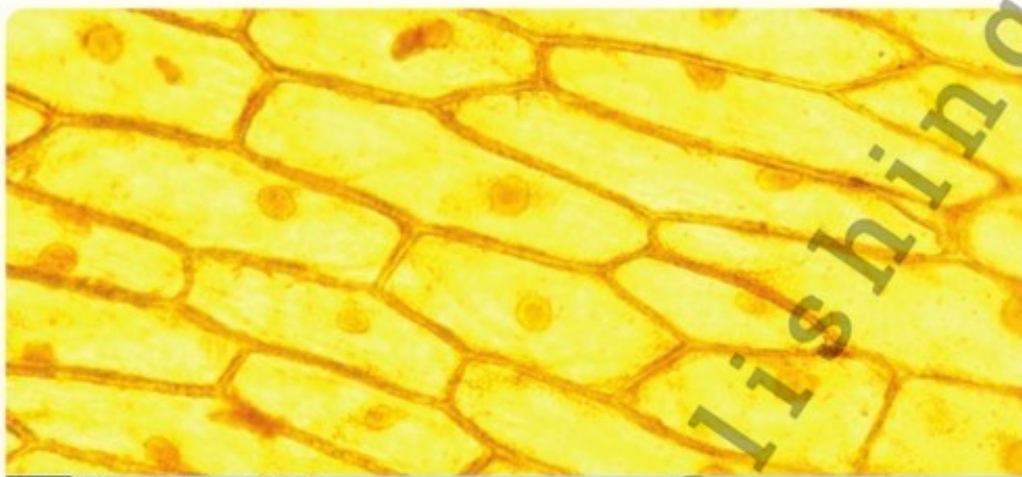


3.17 A eukaryotic cell (TEM): a plant cell showing the pink nucleus surrounded by a double membrane; the nucleolus is brown

Experiment 3.1 To be familiar with and to use a light microscope

- 1 Make sure that the lenses are clean.
- 2 Lower the microscope stage as far as it will go.
- 3 Click the low-power objective lens into place.
- 4 Place a microscope slide on the stage. Ensure that the object to be viewed is in the centre of the opening in the stage.
- 5 Clip the slide into position.
- 6 View the stage from the side and use the coarse adjustment knob to move the low-power objective lens down so that it is just above the slide.
- 7 View the object through the microscope.
- 8 Adjust the coarse adjustment knob to move the stage down until the object is in focus. (Steps 6–8 prevent the slide from being damaged by the objective lens.)
- 9 Adjust the amount of light so that the object can be seen most clearly (this often involves reducing the amount of light). Depending on the type of microscope being used this may involve one or more of the following procedures:
 - ▶ Adjusting the condenser to focus light on the object
 - ▶ Adjusting the diaphragm to control the amount of light reaching the object
 - ▶ Adjusting the angle of the mirror
 - ▶ Using the concave side of the mirror
 - ▶ Placing a sheet of paper between the bulb and the microscope to cause the light to be diffused.
- 10 When the object is focused under low power, move the slide so that the part of the object you wish to view is in the centre of what you can see (called your field of view).
- 11 Click the high-power objective lens into place.
- 12 The object should be almost in focus. If it is not, use the fine adjustment knob to focus it correctly. Be careful not to move the objective lens too close to the slide (as this would crack the slide).

Experiment 3.2a To prepare and examine plant (onion) cells, using a light microscope



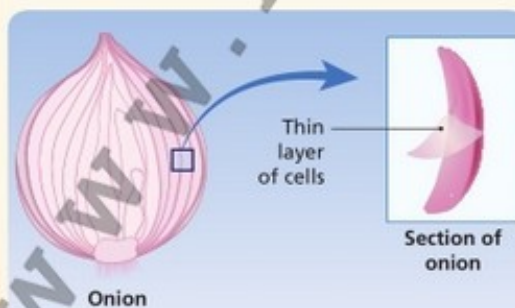
3.18 Onion cells stained with iodine

A Prepare the slide

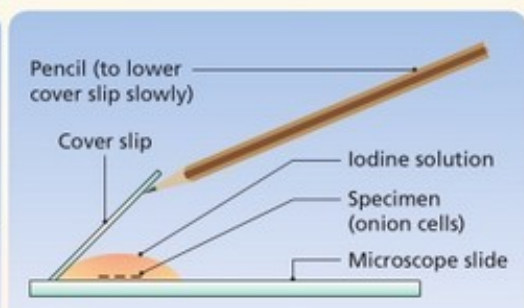
- 1 Remove the outer, dry scaly leaves of an onion.
- 2 Use a forceps or your fingers to pull a strip of thin, transparent epidermis from the inner curve of a fleshy, inner leaf.
- 3 Place a small piece of the epidermal strip on a microscope slide.
- 4 Add a few drops of iodine solution. (This is a red-yellow stain. It stains the nucleus orange and the cytoplasm yellow. A mixture of potassium iodide and iodine gives a better result.)
- 5 Add a cover slip (this prevents the cells from drying out and prevents the lens from getting stained). Lower the cover slip at an angle (this eliminates air bubbles).
- 6 Blot off any surplus iodine, if necessary.
- 7 The cells can be viewed unstained by using a few drops of water instead of iodine solution at step 4 above.

B Examine under the microscope

- 1 The slide can be examined under the microscope in the same way as described in Experiment 3.1.
- 2 The results will appear similar to those shown in diagram 3.18.
- 3 Draw diagrams of what you can see at low power and at high power.

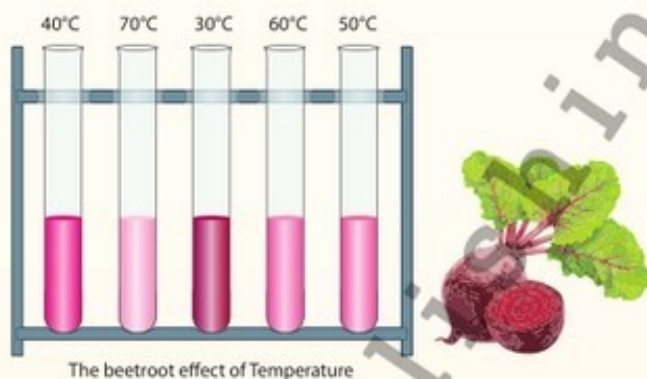


3.19 Obtaining onion cells



3.20 Lowering the cover slip

Experiment 3.2b To investigate the effect of Temperature on the permeability of cell membranes



3.21 Test for investigating the effect of temperature on the permeability of cell membranes

Method

- 1 Cut the beetroot cylinders into 5 x 1cm segments. Place in a strainer and wash off any pigment that has leaked out due to the cutting process.
- 2 Label 5 small test tubes: 70, 60, 50, 40, 30°C.
- 3 Add one piece of beetroot to each of the test tubes.
- 4 Add 3ml of water to each test tube and place in water baths heated to 70, 60, 50, 40, 30°C. Leave for 15 minutes – shake occasionally
- 5 After 15 minutes transfer the content of the test tubes in the water bath to respective empty test tubes also labelled: 70, 60, 50, 40, 30°C.
- 6 Remove the beetroot segments and place in the bin.
- 7 Record the transmittance of each of the coloured solutions at 565 nm. Ensure that you blank the colorimeter before taking the readings. Suggest an uncertainty for the calorimeter.

Conclusions

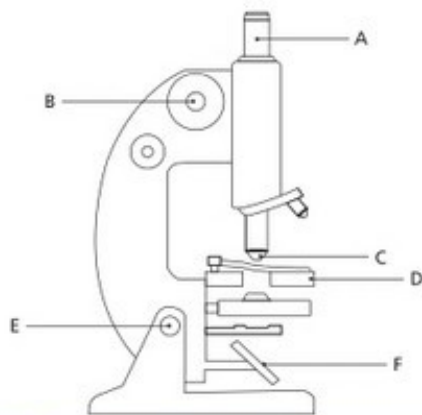
What conclusions can you draw from the experiment? How can you link these observations to membrane structure?

Consider:

- ▶ proteins
- ▶ bonds holding polypeptide chains
- ▶ temperature
- ▶ unfolding of polypeptide
- ▶ denaturation
- ▶ leakage of pigment
- ▶ absorbance of light

Questions on Module 3

- 1 (a) Name the parts labelled A, B, C, D, E and F on the diagram of the microscope below.



3.22 Compound light microscope

- (b) Give the reason why this is an example of a compound microscope.
- (c) What are the functions of the parts labelled B and C?
- (d) If lens A is marked $\times 10$ and lens C is marked $\times 20$, what is the total magnification of the image?
- 2 When using microscopes explain why:
- Thin specimens are used
 - It can be advantageous to place the specimen in water
 - Stains are sometimes used
 - You should focus the objective lens upwards (away from the slide) first
 - You use low power first, followed by high power
 - A cover slip is used
 - The cover slip is placed on the slide at an angle.
- 3 (a) Name any stain you have used for microscopic examination.
- (b) What was the stain used to highlight?
- (c) What colour did the stain appear under the microscope?
- 4 (a) Suggest any advantage of an electron microscope compared with a light microscope.
- (b) What is meant by cell ultrastructure?
- 5 (a) In what cell organelle are amino acids joined together?
- (b) What is formed in these organelles?
- 6 Name the material(s) that form the: (a) Plant cell wall (b) Nucleus (c) Cell membrane.
- 7 (a) Give the functions of: (i) Cell surface membrane (ii) Cell wall (iii) Nucleus (iv) Vacuoles (v) Mitochondria (vi) Chloroplasts (vii) Ribosomes.
- (b) Which of the structures named in part (a) are found only in plants?
- 8 (a) Draw a labelled diagram of an animal cell as seen using the light microscope.
- (b) What extra structures might be seen if a plant cell were drawn?
- 9 (a) Draw a diagram to show the ultrastructure of an animal cell. Label at least six parts.
- (b) Repeat part (a) for a plant cell.
- 10 (a) Why do sperm cells have many mitochondria?
- (b) Name a type of animal cell that has few mitochondria.
- 11 Distinguish between prokaryotic and eukaryotic cells on the basis of:
- Nucleus
 - Cell organelles
 - Size
- 12 Light microscopes are much cheaper than electron microscopes. Give two other advantages of a light microscope over an electron microscope.
- 13 A light microscope has a circular field of view. The diameter of the field of view is 2 mm. Calculate the area of the field of view in mm^2 . Give your answer to two decimal places.
- 14 What happens to the area of the field of view in a light microscope when the magnification is increased? Explain your answer.
- 15 Normal laboratory rules must be followed when experimenting with heat. State and explain one other safety precaution needed when carrying out temperature experiments.
- 16 Give two similarities and two differences between mitochondria and chloroplasts.
- 17 Which of these structures are not surrounded by a membrane? Give the numbers of any that apply.
- vacuoles
 - chloroplasts
 - mitochondria
 - ribosomes
 - nucleus

Module 4 Transport in the Blood

Learning objectives

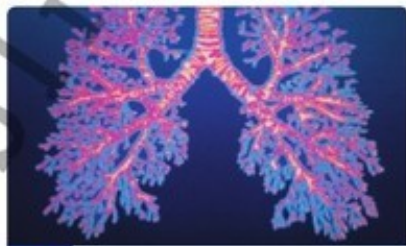
- Explain oxygen dissociation curves for adult and foetal haemoglobin and myoglobin (10.1.3.1)
- Calculate surface area to volume ratios and explain their significance in relation to the transport of materials (10.1.3.2)
- Explain the mechanism of passive transport (10.1.3.3)

Lungs

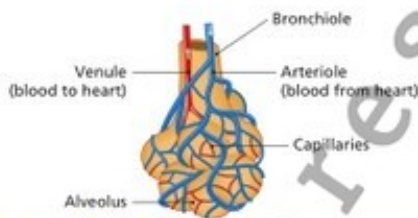
The lungs are large, pink, spongy structures in which gas exchange takes place. Each lung is enclosed by a pair of pleural membranes (the pleura):

- The outer pleura lines the chest wall and the diaphragm
- The inner pleura lines the lungs.

The pleural cavity is the gap between the two pleura. It contains a liquid which lubricates the membranes and reduces friction during breathing.



4.1 Computerised tomography (CT) scan of lungs



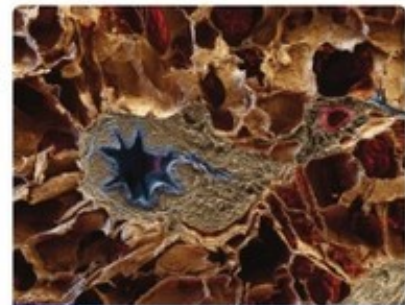
4.2 Bronchiole and alveoli

Alveoli

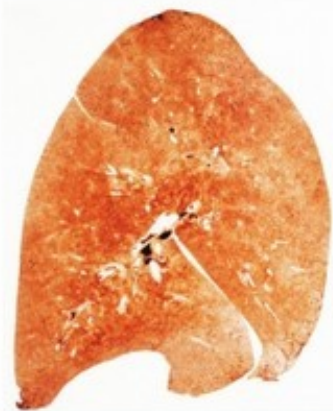
Each bronchus subdivides into about one million bronchioles. These end in tiny, hollow, balloon-like air sacs called alveoli (singular alveolus).

The function of the alveoli is gas exchange. They are adapted for this function because:

- The huge number of alveoli (over 700 million between the two lungs) provide a huge surface area for gas exchange
- They are thin walled (only one cell thick)
- They have moist surfaces
- They are enclosed in a network of blood capillaries.



4.3 Section of lung: alveoli (brown), a bronchiole (blue) and a blood vessel (red)



4.4 A healthy lung



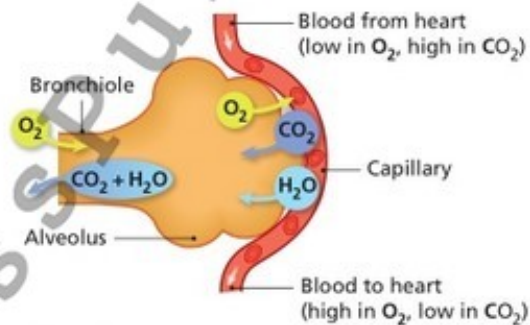
4.5 A smoker's lung, showing tar deposits

Gas exchange

Respiration takes place in body cells in order to supply the cells with energy. As a result body cells use up oxygen and produce carbon dioxide and water.

Carbon dioxide and water

Carbon dioxide and water pass out of body cells by diffusion. This happens because the cytoplasm has a higher concentration of carbon dioxide and water than the blood plasma. In the lungs, carbon dioxide and water diffuse from the blood plasma into the alveoli (i.e. from a high to a lower concentration).



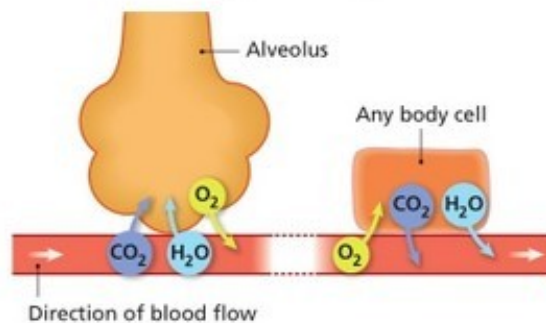
4.6 Gas exchange in an alveolus

Oxygen

Oxygen diffuses in the reverse direction. It passes from the alveoli into the blood and then it passes from the blood into the body cells. In each case it is diffusing from a higher to a lower concentration.

Transport of gases

- Oxygen is mainly transported by combining with the red pigment haemoglobin, to form oxyhaemoglobin. Only about 3% of oxygen is carried dissolved in plasma.
- Carbon dioxide and water are both carried in blood plasma.



4.7 Gas exchange in an alveolus and a body cell

Red blood cells

Red blood cells (also called red blood corpuscles or erythrocytes) are produced in the marrow of bones such as the ribs, breastbone, the long bones in the arms and legs, and the vertebrae of the backbone.

Red blood cells are round and very small – about 5 million are found in 1 cubic millimetre (mm³) of blood. They consist of a flexible membrane containing many molecules of a red protein called haemoglobin.

Red blood cells are broken down (and replaced) at the amazing rate of 3 million cells per second. This is because they become damaged by constantly changing shape in order to pass through narrow blood vessels.

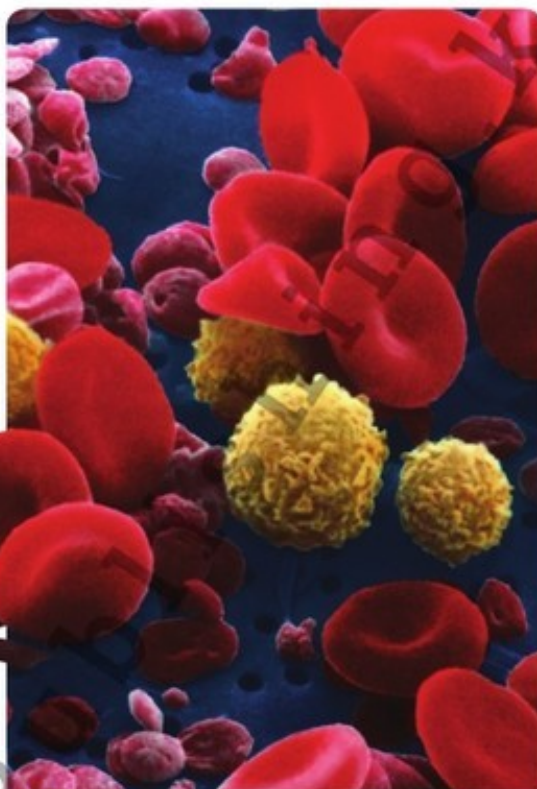
Red blood cells cannot repair themselves and so they live for only about 4 months. Dead red blood cells are broken down in the liver and spleen.

- The iron from the haemoglobin is stored in the liver and may be recycled to make new haemoglobin in bone marrow.
- The rest of the red blood cell and haemoglobin is converted to bile pigments such as biliverdin and bilirubin.

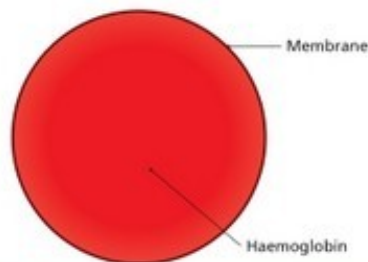
Role of red blood cells

The role (or function) of red blood cells is to transport oxygen.

Haemoglobin is based on molecules of iron and can join with oxygen in areas of high oxygen concentration (e.g. the lungs) and release oxygen in areas of low oxygen (e.g. the body cells).



4.8 Red blood cells (red), white blood cells (yellow) and platelets (pink)



4.9 A red blood cell



Anaemia is a lack of haemoglobin (or red blood cells). The symptoms of anaemia are pale skin colour and a loss of energy.

Features of red blood cells

Red blood cells have:

- No nuclei (when mature)
- No mitochondria
- Biconcave shape (to give a larger surface area).

When red blood cells (erythrocytes) are first made they have a nucleus. They lose their nuclei within a few days, so mature red blood cells have no nuclei. They are then called red blood corpuscles. They also lack mitochondria.

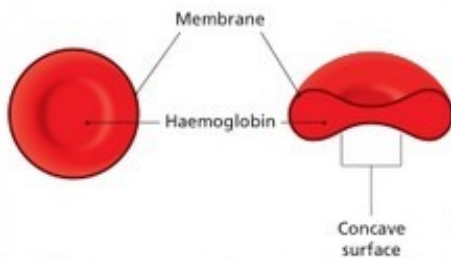
Red blood cells are said to have a biconcave shape. This gives them a larger surface area over which they can exchange oxygen.

Haemoglobin has an amazing ability to form a loose chemical union with oxygen. In the lungs haemoglobin combines with oxygen to form oxyhaemoglobin. Haemoglobin is a purple colour; oxyhaemoglobin is a bright red colour.

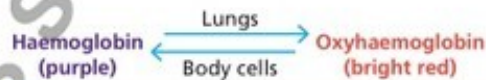
Fortunately, haemoglobin loses oxygen very readily, which allows it to supply the cells in the body with oxygen.



4.10 Biconcave red blood cells (SEM)



4.11 Biconcave shape of a red blood cell



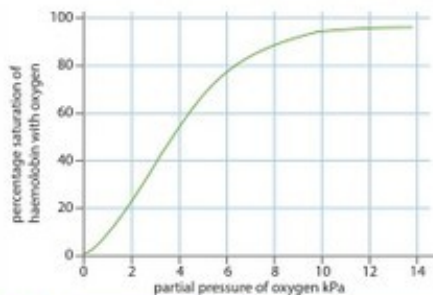
4.12 Relationship between haemoglobin and oxyhaemoglobin

Dissociation Curve

A graph of the percentage saturation of blood with oxygen, [the amount of oxyhaemoglobin compared to haemoglobin at different partial pressures] is shown in Fig. 4.13. This form of graph is called an oxygen dissociation curve:

In the lungs, where the concentration of oxygen is high, haemoglobin will take up oxygen and form oxyhaemoglobin. In the tissues where the oxygen concentration is low, the oxyhaemoglobin will dissociate and release the oxygen.

At high partial pressures of oxygen, haemoglobin has a high affinity (attraction) for oxygen and is highly saturated. At low partial pressures, the affinity is lower and the oxyhaemoglobin dissociates and is less saturated. The curve is steep because a small change in partial pressure causes a massive loading or unloading of oxygen.



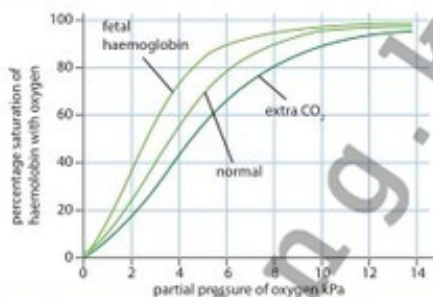
4.13 Dissociation Curve

Changes to the dissociation curve

Several different factors can cause changes to the dissociation curve as shown in Fig. 4.14

Fetal haemoglobin has a greater affinity for oxygen than adult haemoglobin which allows the fetus to take oxygen from the mother's haemoglobin. This is represented by the steeper curve in Fig. 4.14

Higher amounts of carbon dioxide at the tissues will cause more oxygen to disassociate as carbon dioxide lowers the affinity of haemoglobin for oxygen and this is represented in the graph where the curve moves to the right. This is called the Bohr effect. The amount of O_2 carried and released by Hb depends not only on the partial pressure of oxygen but also on pH. An acidic environment causes HbO_2 to dissociate and release the O_2 to the tissues. Just a small decrease in the pH, from the presence of increased levels of carbon dioxide, results in a large decrease in the percentage saturation of the blood with O_2 .



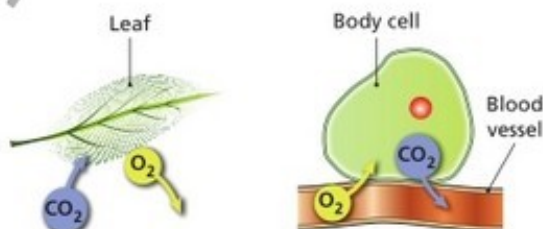
4.14 Adult and fetal dissociation curves

Myoglobin

Myoglobin is another substance that can act as a respiratory pigment. It does not travel in the blood but is found in muscle. It has a greater affinity for oxygen than haemoglobin and so only releases oxygen at very low partial pressures. It acts as a store of oxygen trying to prevent anaerobic respiration from occurring.

Diffusion

Diffusion is said to take place along a concentration gradient. Everyday examples of diffusion include the way the smell of perfume, bread baking or the unpleasant effects of a stink bomb can spread through a room.



4.15 Examples of diffusion

Diffusion is the spreading out of molecules from a region of high concentration to a region of low concentration.

Examples of diffusion in biology are carbon dioxide diffusing into a leaf, oxygen diffusing out of a leaf, oxygen diffusing from the blood into a cell and carbon dioxide diffusing out of a cell.

Diffusion is caused by the kinetic energy of the molecules. These molecules are moving randomly and will tend to spread out if they can. This movement does not need external energy so diffusion is said to be passive.

4.16 Diffusion: the dark blue solution is spreading out into the pale blue solution



Experiment To demonstrate the effect of surface area to volume ratio on diffusion

Materials:

- Pre-prepared agar-agar
- A ruler
- 0.1M NaOH per group 200 mL
- Knife
- 400ml beaker
- Tongs



4.17 Agar-agar cubes

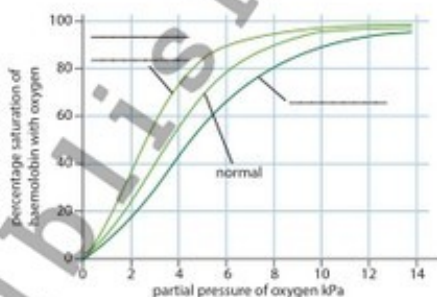
Procedure:

- 1 Cut three cubes of prepared agar-agar: A 3cm cube, a 2cm cube, and a 1cm cube.
- 2 Pour 200mL of 0.1M sodium hydroxide solution into the beaker.
- 3 Immerse the 3 cubes in the sodium hydroxide solution, noting the time.
- 4 Let the cubes soak for approximately 10 minutes.
- 5 Gently stir the solution every few minutes.
- 6 After 10 minutes, use a spoon or tongs to remove the cubes from the sodium hydroxide solution.
- 7 Blot the cubes with a paper towel.
- 8 Cut each cube in half and measure the depth to which the colour has penetrated. Sketch the cross-section of each block.
- 9 Record your measurements and sketch each cross-section of the cube:
 - ▶ Calculate total volume of each cube (volume = $L \times W \times H$)
 - ▶ Calculate volume that did not turn pink. (Calculate total volume of the small portion of the cube that does not change colour – use the same formula $L \times W \times H$)
 - ▶ Calculate volume diffused = total volume – volume not change colour
 - ▶ Calculate % diffusion = $\text{Volume diffused} / \text{total volume} \times 100$

Questions on Module 4

- In terms of maximizing diffusion, what was the most effective size cube that you tested?
- Why was that size most effective at maximizing diffusion? What are the important factors that affect how materials diffuse into cells or tissues?
- If a large surface area is helpful to cells, why do cells not grow to be very large?
- In what ways does the body adapt surface area-to-volume ratios to help exchange gases?
- Explain the difference between a typical adult and foetal oxygen dissociation curve?
- Why is the oxygen dissociation curve (ODC) S shaped?
- What is the Bohr effect?
- Name the process involved in the passage of gas between the alveolus and the blood.
- What are the functions of alveoli?
- 'Gas exchange in the alveoli is the reverse of gas exchange in muscle cells.' With reference to two named gases explain why this statement is true.
- State a precise location in the human body at which red blood cells are made.
- What shape are red blood cells?
- What is the advantage of this shape?
- Suggest two reasons why red blood cells have short lives.
- Where are red blood cells broken down?
- Name the end product made from the breakdown of red blood cells.
- What is diffusion?
- In the case of a named molecule, give a precise location at which it diffuses in the human body.
- What causes anaemia?
- What are the symptoms of anaemia?
- When we breathe in, our alveoli fill with air that contains about 21% oxygen. When we breathe out, the air contains about 14% oxygen. Explain why the percentage of oxygen in air we breathe out does not drop to 0%.

- Myoglobin is a protein found in muscles that binds oxygen. It has a greater affinity for oxygen than haemoglobin.
 - Explain why myoglobin needs to have a greater affinity for oxygen than haemoglobin.
 - The graph shows the oxygen dissociation curve for haemoglobin. Label the gaps.



4.18 Dissociation curves

- When a molecule of oxygen diffuses from the air in alveoli into a red blood cell, what is the minimum number of cell surface membranes it must pass through?
- Muscle cells that are active release both carbon dioxide and water into the blood. What process in the muscle cells releases water?
- At the lungs, most carbon dioxide diffuses from the capillaries into the air in alveoli. Explain why not all carbon dioxide molecules diffuse in this direction.
- The rate of diffusion of oxygen and carbon dioxide is related to the rate of respiration in cells. It is also related to the rate of photosynthesis in plant cells. During the day, the net movement of oxygen is out of plants, and the net movement of carbon dioxide is into plants. At some point in the evening and again in the morning, there is no net diffusion of gases into or out of a living plant. What causes this?

Module 5 Respiration

Learning objectives

- Describe the structure and function of ATP (10.1.4.1)
- Compare ATP synthase in aerobic and anaerobic respiration (10.1.4.2)
- Name the types of metabolism (10.1.4.3)
- Describe the stages of energy exchange (10.1.4.4)
- Establish the relationship between mitochondrial structure and cellular respiration processes (10.1.4.5)
- Describe the Krebs cycle (10.1.4.6)

Introduction

Gas exchange is the process by which organisms exchange gases with their environment. Gas exchange takes place in the lungs of mammals, through the gills in fish, through openings in plant stems and on the bottom surface of leaves.

Respiration is the controlled release of energy from food. The food involved is usually glucose.

The process of respiration is controlled by enzymes. They allow energy to be released in small amounts, which can easily be trapped for later use. The energy is trapped in the form of a molecule called ATP (adenosine triphosphate).

Respiration can be either aerobic or anaerobic.

Aerobic respiration

Most living things get their energy from aerobic respiration and are therefore called aerobes. In this process, the energy stored in the bonds of a molecule such as glucose is released and used to make ATP.

When ATP later breaks down, it supplies energy for all the metabolic reactions in the cell (such as muscular movement, production of new cells and growth).

Aerobic respiration is a relatively efficient method of obtaining energy. About 40% of the energy in glucose is converted to ATP during aerobic respiration. This high efficiency is due to the substrate (glucose) being completely broken down.

The end products of aerobic respiration (carbon dioxide and water) have very low energies. Most of the energy that is not converted to ATP is lost as heat.

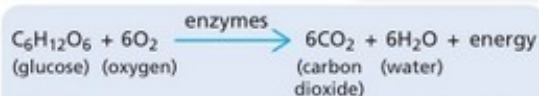


5.1 Respiration produces energy



5.2 Summary of respiration

Aerobic respiration is the controlled release of energy from food using oxygen.



5.3 A balanced equation to represent aerobic respiration

ADP and ATP

ADP

ADP is the abbreviation for **adenosine diphosphate**. ADP is found in the cells of all organisms.

ADP is made of the base adenine (this base is also found in DNA and RNA), a five-carbon sugar called ribose and two phosphate groups. The bond between the two phosphate groups is an unstable bond. These unstable bonds are represented by the symbol '-'.

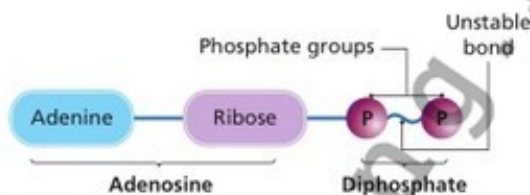
ADP is a low-energy molecule.

ATP

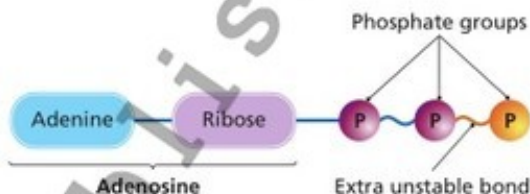
ATP is an abbreviation for **adenosine triphosphate**. ATP forms when another phosphate group is added to ADP. Extra energy is also added, in the form of the unstable bond between the last two phosphate groups.

The process of adding a phosphate group is called **phosphorylation**.

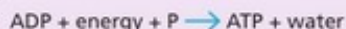
ATP is an energy-rich compound. It can be moved around inside a cell, i.e. it is an energy carrier. However, it cannot be stored for very long. The energy in ATP must be used immediately.



5.4 The structure of ADP



5.6 The structure of ATP



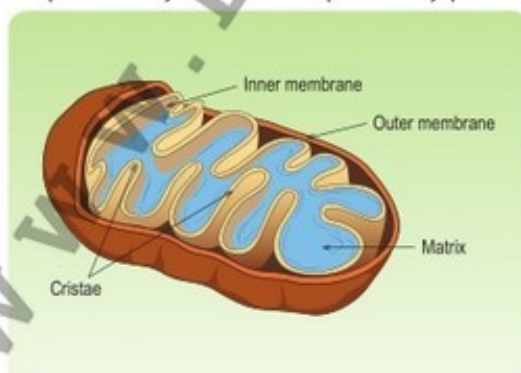
5.7 The formation of ATP



5.8 The breakdown of ATP

Mitochondria

Mitochondria are elongated organelles with a double membrane structure. The inner membrane is folded inside to form Cristae [see diagram below] which are folded in the Matrix - the central part of a Mitochondrion. The most important function of the mitochondria is to produce energy. The simpler molecules of nutrition are sent to the mitochondria to be processed and to produce charged molecules. These charged molecules combine with oxygen and produce ATP molecules. This process is known as oxidative phosphorylation. The folds of the inner membrane enhance the productivity of cellular respiration by providing the maximum surface to area ratio for diffusion.



5.9 Mitochondria



5.10 Mitochondria viewed through an electron microscope

Stages in aerobic respiration

Respiration may occur as either a one-stage or a two-stage process. Aerobic respiration is a two-stage process.

Stage 1

- Is an anaerobic process (this means it does not use or require oxygen in order to take place).
- Releases only a small amount of energy – as a result, it is inefficient as an energy-release system.
- Takes place in the cytosol of the cell. The cytoplasm consists of all of the living parts of the cell surrounding the nucleus. The cytosol is the cytoplasm minus the cell organelles.
- Involves splitting glucose (a six-carbon sugar) into two three-carbon molecules. In doing this, a small amount of energy is released and used to produce a small number of ATP molecules.

In stage 1, glucose is incompletely or partially broken down. Much of the energy that was stored in the bonds of the glucose molecule is retained in the bonds of the two three-carbon molecules. This is why stage 1 produces only a low yield of energy.

Stage 2

- Is an aerobic process (it requires and uses oxygen).
- Releases a large amount of energy. This means it is efficient as an energy-release system.
- Takes place in the mitochondria.

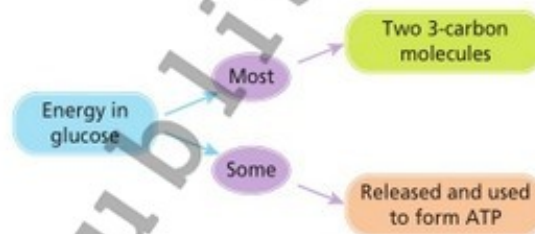
The stage 2 reactions are very complex. They involve the breakdown of the three-carbon molecules (formed in stage 1) to carbon dioxide and water. One of the steps in this breakdown requires oxygen.

As stage 2 is a complete breakdown of the three-carbon molecules, it releases a large amount of energy (which is used to form a large number of ATP molecules). Very little energy remains in the carbon dioxide and water molecules.

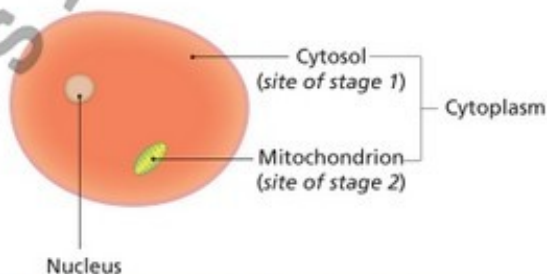
Carbon dioxide and water are the end products of both stage 2 and of aerobic respiration.

Glucose → two 3-carbon molecules + a small amount of energy

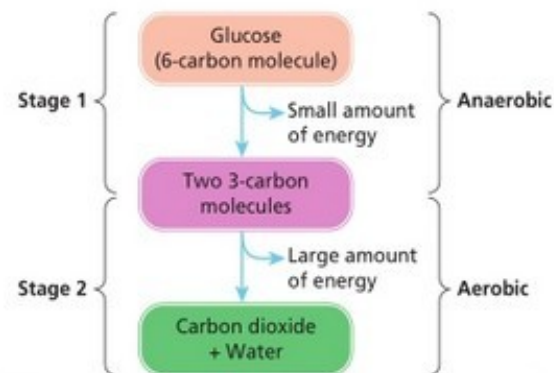
5.11 A word equation to represent stage 1 of respiration



5.12 Energy in stage 1 of respiration



5.13 Location in a cell of stages 1 and 2 of respiration



5.14 Respiration is a two-stage process

Anaerobic respiration

Anaerobic respiration:

- Is also known as fermentation
- Does not use oxygen (note it can occur in the presence of oxygen, but it does not use oxygen)
- Is a stage 1 process (i.e. it does **not** involve stage 2)
- Takes place in the cytosol
- Releases a small amount of energy
- Initially involves the breakdown of glucose into two three-carbon molecules.

Anaerobic respiration is the controlled release of energy from food without the use of oxygen.

There are many different forms of anaerobic respiration. In each of them, the three-carbon molecules are converted to some other end products, but no extra energy is released.

Anaerobic respiration is a far less efficient process than aerobic respiration, because glucose is only partially broken down.

Two common types of fermentation are:

- Lactic acid fermentation (often simply called anaerobic respiration)
- Alcohol fermentation.

Fermentation is another name for anaerobic respiration.

Lactic acid fermentation

Lactic acid fermentation occurs in some bacteria and fungi (which are called anaerobes) and in human muscle when it is short of oxygen. In this process the three-carbon molecules are converted to lactic acid.

Lactic acid fermentation takes place in human muscle when the supply of oxygen to the muscle is not sufficient to meet its energy needs (i.e. when we are out of breath). Lactic acid builds up in the muscle, causing cramp and muscular stiffness. When the person rests, the lactic acid is taken to the liver by the blood and broken down.

Lactic acid (which can easily form a similar substance called lactate) is formed when:

- Bacteria cause milk to go sour
- Bacteria respire on cabbage to produce sauerkraut
- Silage is made
- Bacteria act on dairy products to make cheese and yoghurt.

Glucose \rightarrow 2 lactic acid + a small amount of energy

5.15 A word equation to represent lactic acid fermentation



5.16 Treating cramp in a calf muscle

Alcohol fermentation

Alcohol fermentation is another form of anaerobic respiration. It takes place in some bacteria, in fungi (such as yeast) and in plants when they are deprived of oxygen.

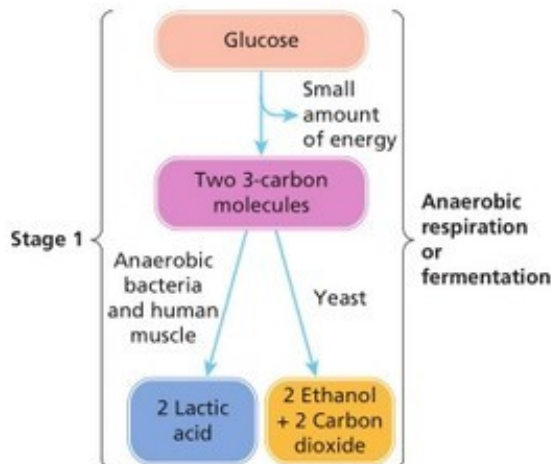
In this process the three-carbon molecules are converted to ethanol and carbon dioxide. This is also a partial breakdown of glucose. The end products of alcohol fermentation are ethanol and carbon dioxide. Ethanol is a high-energy product.

Glucose \rightarrow 2 ethanol + 2 carbon dioxide + a small amount of energy

5.17 A word equation to represent alcohol fermentation



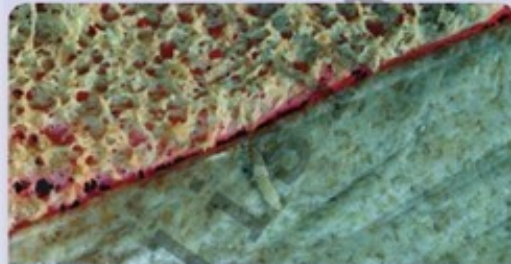
5.18 Yeast cells (scanning electron micrograph)



5.19 The two types of anaerobic respiration



Alcohol fermentation has been used for thousands of years in baking and in beer and wine production. In baking, special yeasts are mixed with flour and liquid to form dough. Alcohol fermentation occurs in the dough. The alcohol evaporates, but the carbon dioxide produced causes the dough to rise, forming a lighter bread.



5.20 Dough rising: unrisen dough is in the bottom and rising dough is shown above

In modern baking, yeast (which cannot withstand the high temperatures of an oven) is often replaced with baking powder as a source of carbon dioxide.



Strictly speaking, 'fermentation' means anaerobic respiration. However, in the bioprocessing industry, fermentation is taken to mean the growth of microorganisms in liquid under any condition (i.e. it can be aerobic or anaerobic).

Microorganisms in industrial fermentation



Biotechnology is the use of living things or their components (especially cells and enzymes) to manufacture useful products or to carry out useful reactions.

Biotechnology can use microorganisms, plants or animals to manufacture products. However, microorganisms are the basis of most production techniques.

Fermentation is the foundation of much of the biotechnology industry. The production of substances using fermentation techniques is a form of bioprocessing.

Production method

In bioprocessing, the microorganisms are placed in a container along with a suitable substrate on which they can react. The vessel in which the biological reactions take place is called a **bioreactor**.

Many different bioreactors are used, but a typical example is shown in diagram 5.21.

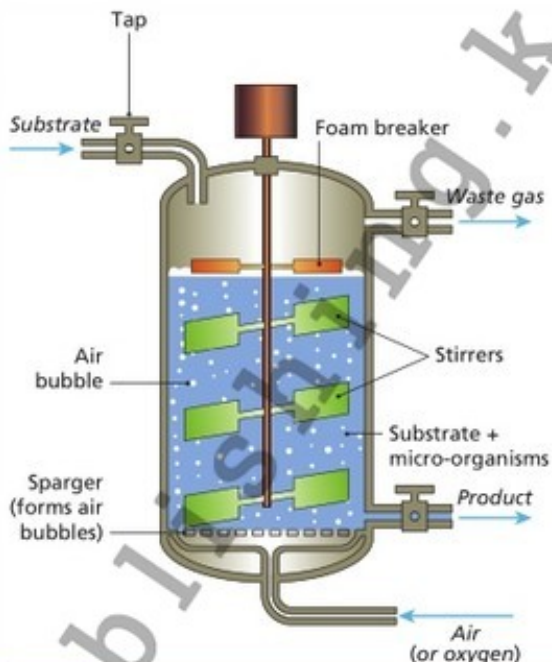
The contents of the bioreactor are mixed so that the microorganisms are brought into contact with the substrate. Sometimes the mixing (or the reaction itself) produces foam. This is reduced by the use of a foam breaker.

The culture medium is the liquid (including a suitable substrate) in which the microorganisms grow. In many bioreactors it is important to get as much oxygen dissolved in the culture medium as possible. For this reason air (or oxygen) is pumped into the vessel. The air passes through a device called a sparger, which forms small air bubbles that dissolve more readily into the culture medium.



5.21 Vaccines being produced in bioreactors

Apart from using the correct microorganism and substrate, the quality and yield of the product depend on a number of other factors. These include the materials and design of the bioreactor, elimination of microorganisms that would contaminate the process, the correct rate of mixing, and control of environmental factors such as temperature and pH.



5.22 A bioreactor

Microorganisms used in bioprocessing

There is a very wide range of microorganisms used in industrial fermentations. New microorganisms are continuously being developed and used. Many of these are produced using genetic engineering techniques.

In general, the main organisms used are bacteria and fungi, especially different yeast strains. Note that very often different organisms may be used to produce the same end product (e.g. antibiotics are produced by bacteria, fungi and yeast).

Very old examples of bioprocessing include the use of yeast in the production of alcohol and of carbon dioxide in baking. Some modern examples of bioprocessing are given in the following table.

Microorganism	Product	Use
Bacteria	Ethanol	Beer, wine, paint, perfume, polish
	Acetone	Solvent (e.g. nail varnish remover)
	Amino acids and vitamins	Food additives (e.g. in breakfast cereals)
	Yoghurt	Food
	Methane gas	Fuel (called biogas)
	Antibiotics	To kill other bacteria
	Enzymes	Washing powders (act as stain removers)
	Drugs	Maintain health
	Hormones	Maintain health
	Yeasts	Ethanol
Carbon dioxide		Causes dough to rise
Single-cell protein		Edible protein
Other fungi	Citric acid	Food and drink additive (e.g. soft drinks)
	Antibiotics	To treat bacterial infections

Bioprocessing with immobilised cells

When microorganisms are used in a bioreactor they are removed, along with the product, at the end of the process. They then have to be separated from the product and new microorganisms must be grown to replace those lost. This is wasteful and costly.

To prevent these problems, microorganisms are often fixed or immobilised in the bioreactor. Whole cells are immobilised in the same way as enzymes.

Detailed study of respiration

Aerobic respiration

Aerobic respiration is a two-stage process (as outlined earlier in this module). We will now look at these stages (especially stage 2) in more detail.

Stage 1

Stage 1 reactions are called glycolysis.

Glycolysis:

- Takes place in the cytosol of the cell
- Does not require oxygen (it is anaerobic)
- Forms two ATP molecules
- Produces pyruvic acid as an end product (pyruvic acid can easily form a charged molecule called pyruvate).

Stage 2

In stage 2 a complex series of reactions take place, some of which require oxygen. If oxygen is present, pyruvic acid enters a mitochondrion. Here it loses a carbon dioxide molecule to form a two-carbon (C_2) molecule called acetyl coenzyme A (often shortened to acetyl CoA).

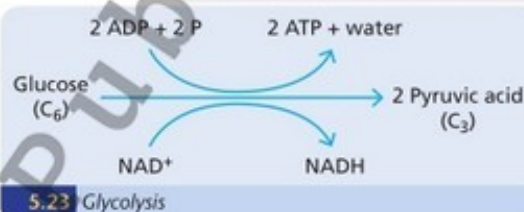
Pyruvic acid also loses two high-energy electrons and a proton (H^+). These combine with NAD^+ to form NADH, as shown in diagram 5.24. Each NADH will enter an electron transport system, as outlined later.

Krebs cycle

Acetyl CoA now enters into a series of reactions called Krebs cycle.

In Krebs cycle, acetyl CoA is broken down to carbon dioxide and protons (H^+) in a number of reactions (see diagram 5.27). The energy that was in acetyl CoA is released in a number of steps in the form of high-energy electrons. These electrons (along with protons, H^+) are picked up by NAD^+ to form NADH. The NADH molecules enter an electron transport system.

Glycolysis is the conversion of glucose into two molecules of pyruvic acid.



5.24 The formation of NADH as a result of the breakdown of pyruvic acid



The reactions in Krebs cycle were first discovered by German biochemist (Sir) Hans Krebs (1900–81) and others in 1937. For his work in this area he shared the Nobel Prize in 1953.

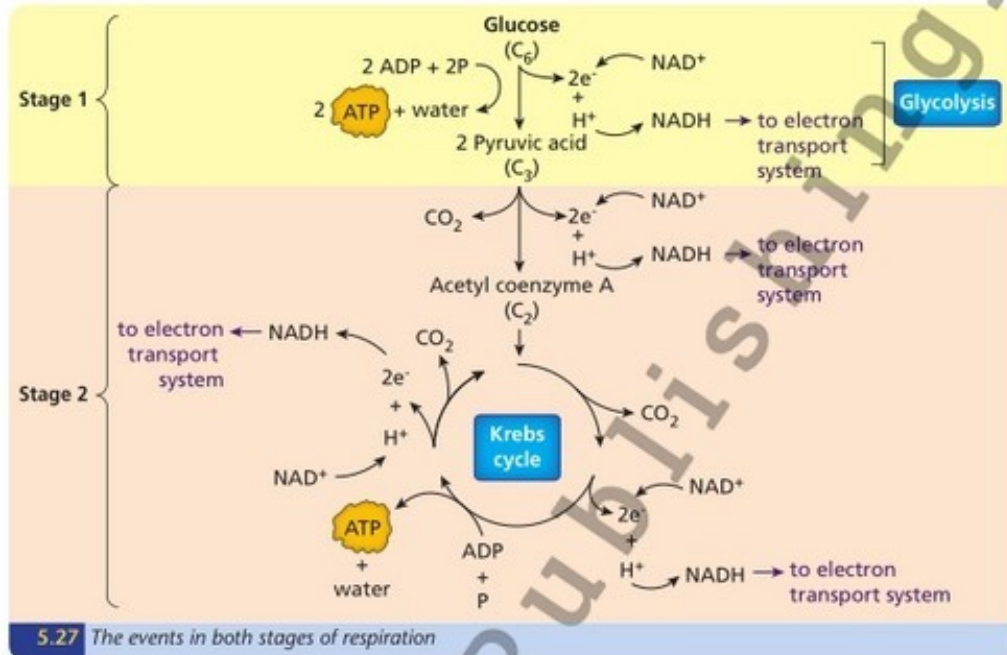


5.25 Sir Hans Krebs

At one point in Krebs cycle a single ADP (and a phosphate) is converted to ATP and water.



5.26 The formation of NADH as a result of the breakdown of acetyl CoA



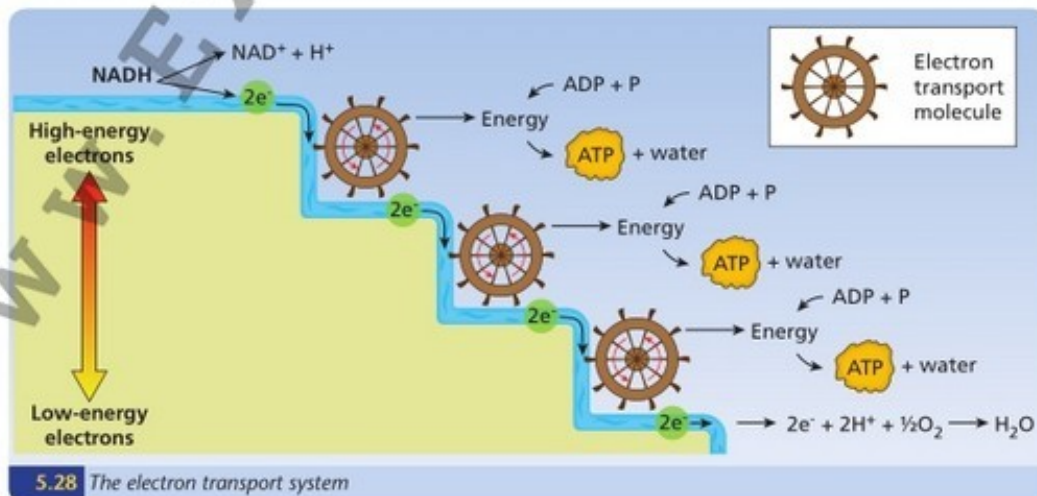
Electron transport system

The electron transport systems, or chains, are located on the inner membrane of the mitochondrion. Infoldings of the inner membrane increase the surface area and allow larger numbers of these systems to fit on the membrane.

Each electron transport system consists of a number of molecules, mostly proteins. High-energy electrons are passed from NADH to the first of these molecules, as shown in diagram 5.28, below.

As the electrons pass from molecule to molecule within each system they lose some of their energy. This is similar to the way water loses energy as it flows down a waterfall. Some of the energy released by the electrons is used to form ATP. The rest of the energy is lost as heat.

At the end of each system, low-energy electrons are removed by combining them with oxygen and hydrogen (H⁺) to form water.





Certain chemicals, such as cyanide, are fatal because they prevent some of the proteins in the system from receiving or passing on electrons. This means that ATP is not produced.

Importance of the electron transport system

The importance or significance of the electron transport system is that it produces the energy-rich carrier ATP. It converts the energy in NADH to a more usable form of energy (ATP).

The electron transport system cannot work in the absence of oxygen. This is because oxygen is essential at the end of the system to remove the low-energy electrons. If oxygen is absent electrons cannot flow along the system and so no further ATP is produced. Aerobic organisms die due to a lack of available energy (ATP), in the absence of oxygen.

Summary of aerobic respiration

Aerobic respiration involves stage 1 and stage 2 reactions (see diagram 5.27).

- **Stage 1** (glycolysis) is anaerobic and releases very little energy.
- **Stage 2** includes Krebs cycle reactions and the electron transport systems. These are aerobic and release a large amount of energy as a result of the complete breakdown of glucose.

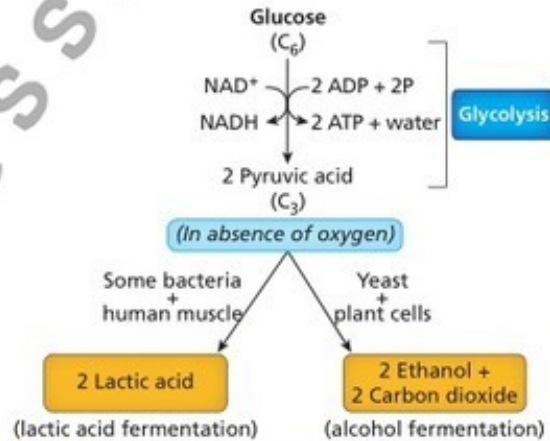
Anaerobic respiration

Anaerobic respiration starts with the stage 1 (glycolysis) reactions. This means that glucose is converted into two molecules of pyruvic acid. ATP and NADH are produced in this process (see diagram 5.29).

In the absence of oxygen, pyruvic acid is converted (or reduced) to either lactic acid (in animals and some bacteria) or ethanol and carbon dioxide (in plants and yeast). In both these forms of anaerobic respiration (also called fermentation) Krebs cycle cannot proceed and no further ATP is produced.

All the reactions in lactic acid fermentation and in alcohol fermentation are stage 1 reactions. These reactions take place in the cytosol and do not involve Krebs cycle or the electron transport systems.

In conclusion, both types of fermentation only involve stage 1 (glycolysis). Stage 1 is anaerobic and releases a small amount of energy.



5.29 The two types of anaerobic respiration

Experiment 5.1 To prepare and show the production of alcohol by yeast**Preparation of alcohol**

- 1 Prepare a glucose solution by dissolving glucose in water.
- 2 Boil the solution in a conical flask for 5 minutes (this eliminates gases from the solution, forming anaerobic conditions).
- 3 When the solution cools, add some dried yeast.
- 4 Cover the liquid in the flask with oil (this prevents oxygen from re-entering the solution).
- 5 Set up either of the two pieces of apparatus as shown in diagram 5.30.
- 6 The airlock is needed to prevent microorganisms entering and to allow carbon dioxide to pass out.
- 7 Limewater or water may be used in the airlock (limewater turns milky in the presence of carbon dioxide).
- 8 The apparatus is placed in a water bath at 30°C (this is an ideal temperature for the maximum rate of respiration).
- 9 Bubbles of carbon dioxide will be seen in the limewater. Fermentation is complete when the bubbles stop forming (often after a few days).
- 10 As a control, the same apparatus is used without adding any yeast cells (or adding boiled yeast). In this case, no bubbles form and the limewater remains clear.



5.30 Preparing alcohol

To show the production of ethanol (the iodoform test)

- 1 Filter the solution (to remove yeast cells).
- 2 Place some of filtrate into a test tube.
- 3 Add an equal volume of potassium iodide solution. Note this is a colourless solution.
- 4 Add sodium hypochlorite solution (note that the solution turns a brown-orange colour and then becomes colourless).
- 5 Place the test tube in a water bath at 50–60°C for 4 or 5 minutes.
- 6 Remove the test tube and allow it to cool.
- 7 The appearance of pale yellow crystals (of a chemical called iodoform) indicates that ethanol is present.
- 8 As a control use water instead of the filtered solution. Yellow crystals do not form.



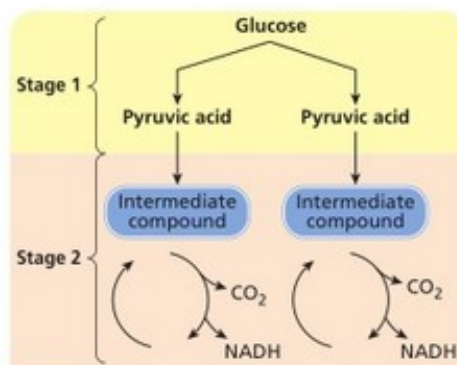
5.31 Yeast fermenting: note the limewater has turned cloudy due to carbon dioxide

To show that alcohol is produced

To test for alcohol, acidified potassium dichromate is added to the filtered solution. The test tube is placed in a warm water bath. If alcohol is present the colour changes from orange to green.

Questions on Module 5

- Distinguish between gas exchange and respiration.
 - State a location for each in humans.
- Why is respiration essential for all living things?
 - Name the normal respiratory substrate.
- Give
 - two similarities and
 - two differences between aerobic and anaerobic respiration.
- Name the end products of:
 - Aerobic respiration
 - Alcohol fermentation
 - Lactic acid fermentation.
- Give a balanced equation for aerobic respiration.
 - Give word equations to represent:
 - Anaerobic respiration in yeast
 - Anaerobic respiration in human muscle.
- Aerobic respiration is a two-stage process. Say whether each of the following statements relates to stage 1 or stage 2.
 - Requires oxygen
 - Takes place in the cytosol
 - Occurs in the mitochondrion
 - Releases a large amount of energy
 - Produces carbon dioxide and water
 - Is a partial breakdown of glucose
 - Is anaerobic.
- Under what conditions do humans respire anaerobically?
 - What is the end product of this process?
 - What is the effect of this product on muscles?
 - State one possible fate of this product in a person.
- What is: (i) Biotechnology (ii) A bioreactor?
 - Name any one organism used in industrial fermentation and state the product of this fermentation.
- Microorganisms are used to produce large quantities of the products listed below. State one commercial application for each of them.
 - Methane gas
 - Enzymes
 - Vitamins
 - Single-cell proteins
 - Citric acid.
- Alcohol fermentation is the oldest known form of industrial fermentation.
 - Name an organism that carries out this process.
 - Name two industries based on this process.
 - State the end product of this process used in each of the industries named.
 - Outline one use for each of the products named in part (c).
- Give a word equation for glycolysis.
 - Name the end products of glycolysis.
 - State the location in a cell for glycolysis.
- State what happens to the products of glycolysis:
 - In the presence of oxygen
 - In the absence of oxygen.
- With regard to Krebs cycle:
 - State its location in a cell.
 - Name the end products.
 - Name the molecule that continuously enters the cycle.
- State the precise location of the electron transport systems in a cell.
- Respiration may be a one or a two-stage process. Distinguish between these stages in terms of:
 - The need for oxygen
 - The amount of energy released
 - Whether one or both occur in
 - aerobic and
 - anaerobic respiration.
- Aerobic respiration can be represented as a two-stage process as shown in diagram 5.25.

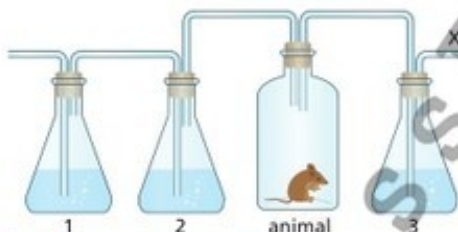


5.22 Aerobic respiration

- Name the two stages shown as 1 and 2.
- Name the intermediate compound shown.
- What is the fate of the
 - carbon dioxide and

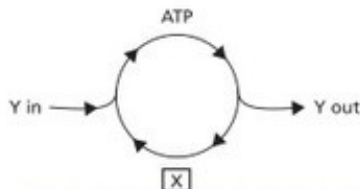
Questions on Module 5

- (ii) NADH, produced at stage 2?
 (d) Name two places on the diagram where ATP is produced.
- 17 The apparatus in diagram 5.33 may be used to demonstrate aerobic respiration. Air is drawn through the apparatus by attaching it to a vacuum pump at X. Sodium hydroxide is placed in flask 1 to remove carbon dioxide.
- What is the purpose of removing carbon dioxide?
 - Limewater is put in flasks 2 and 3. Suggest a reason for putting it in each flask.
 - What is the purpose of a control in an experiment? Suggest a suitable control for this experiment.
 - If the animal in the apparatus were replaced by a plant, and the experiment carried out in daylight, would you expect a similar result? Explain your answer.



5.33 Aerobic respiration experiment

- 18 Choose which of the options (i), (ii), (iii) or (iv) represents the correct answer in each case below.
- Krebs cycle reactions occur in:
 - Chloroplasts
 - Cytosol
 - Mitochondria
 - Yeast.
 - The largest ATP (energy) output results from:
 - Glycolysis
 - Aerobic respiration
 - Anaerobic respiration
 - Stage 1 reactions.
 - Respiration is essential because it:
 - Releases energy
 - Uses glucose
 - Requires enzymes
 - Involves stage 1.
 - In preparing alcohol, anaerobic conditions are achieved by:
 - Using a water bath
 - An airlock
 - Boiling the substrate solution
 - Using a conical flask.
- (e) During anaerobic respiration, lactic acid may be directly produced from: (i) Glucose (ii) Ethanol (iii) Pyruvic acid (iv) ATP.
- 19 When glucose is being used for aerobic respiration, what can be said about the number of molecules of oxygen used and the number of molecules of carbon dioxide produced?
- 20 Yeast used in bread making produces alcohol and carbon dioxide in the dough. Explain why is there almost no alcohol in bread made with yeast.
- 21 When making bread with yeast, the dough containing the yeast is left in a warm place for some time before being put into the oven. Explain what happens to the dough mixture in the warm place and in the oven and why the bread would not rise if the dough was put straight into the oven.
- 22 The most important end product of respiration for all cells is ATP. What is ATP used for?
- 23 Describe the similarities and differences between anaerobic respiration in yeast cells and in animal cells.
- 24 When lactic acid builds up in muscles it can be very painful. Prolonged build-up of lactic acid can be harmful to cells and tissues. Suggest and explain the effects that lactic acid could have on cells and tissues.
- 25 When growing microorganisms in a bioreactor, a growth medium and some other substances must be provided. Name the four chemical elements that are essential for all microorganisms.
- 26 ATP plays an important role in cell activities.
- Name the substance X, formed by the loss of a phosphate group.
 - The ATP cycle is kept going by Y. What is Y?



5.34 The ATP cycle

Module 6 Excretion

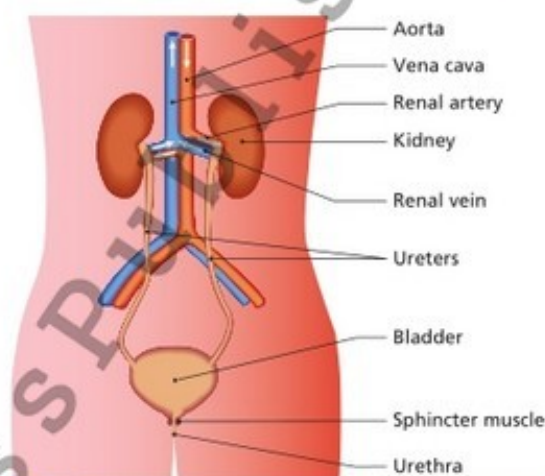
Learning objectives

- Explain the mechanism of filtration and formation of urine (10.1.5.1)
- Explain the role of the antidiuretic hormone (ADH) in water monitoring (10.1.5.2)
- Explain the dialysis mechanism (10.1.5.3)
- Discuss the advantages and disadvantages of kidney transplantation and dialysis (10.1.5.4)

Organs of excretion

The main organs of excretion are:

- **Lungs.** The lungs excrete water and carbon dioxide.
- **Skin.** The skin excretes water and salts (in the form of sweat).
- **Kidneys.** These are the main excretory organs in humans. They excrete water, salts and urea (in the form of urine). By controlling the amount of water and salts that are excreted, the kidneys play a major role in the homeostasis of the fluid and chemical composition of the blood (and of the body).



6.1 The urinary system

The urinary system

Kidneys

The urinary system consists of two kidneys, two ureters, the bladder and the urethra. The kidneys are fist-sized, bean-shaped organs located just below the diaphragm in the small of the back.

Blood in the aorta contains waste products collected from all over the body. Some of this blood enters the kidneys through the two renal arteries. Every minute about 20% of our blood passes into the kidneys.

Main processes in the kidneys

Filtration

In the kidneys the incoming blood is filtered. This takes place in the outer **cortex** of each kidney. Filtration results in small substances (both useful and waste) being forced out of the bloodstream into the kidney.



6.2 Computerized tomography (CT) scan of a section of human kidney

Reabsorption

Some of the useful materials are then taken back into the blood. This is called reabsorption. It occurs in the **cortex** and **medulla** of each kidney.

Secretion

Some substances are secreted from the blood into the **cortex** of the kidney. These substances include potassium and hydrogen ions. (Too much potassium in the body prevents nerve impulses travelling correctly and reduces the strength of muscular contraction.)

By controlling the hydrogen ion concentration of the blood, the kidneys control blood pH. Purified blood leaves the kidneys through the renal veins. The renal veins take the blood to the vena cava.

When these three processes are complete only unwanted waste and toxic products are left in the kidney. These form the liquid called urine.

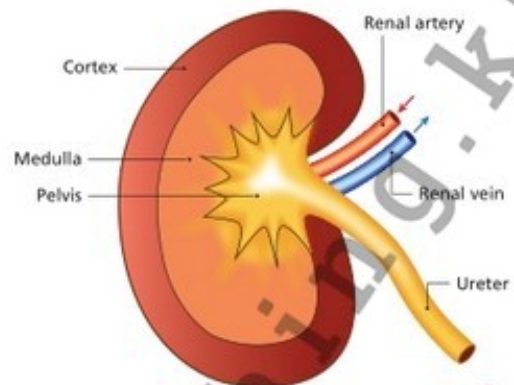
Urine

Urine is typically composed of:

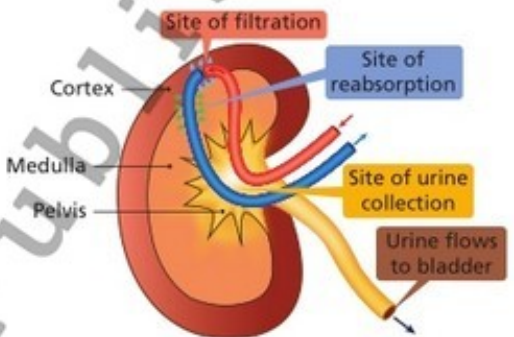
- Water (96%)
- Nitrogenous waste (2.5%, mostly urea)
- Salts (1.5%).



6.5 A human kidney, showing the cortex, medulla and pelvis



6.3 Structure of the kidney



6.4 Simplified diagram to show the location of the main events in urine production

Urea is produced in the liver. It is formed when excess proteins are broken down (de-aminated). Urine flows from the medulla into the renal pelvis. This is shaped like a funnel and collects waste and carries it into the ureter. The waste (called urine) is then carried by the two ureters to the bladder.

Bladder

The bladder stores urine. It is a muscular organ that is not under voluntary control. Two sphincter muscles are located at the junction of the bladder

and urethra. In babies, these muscles open automatically by reflex action when the bladder becomes about half full. The bladder then contracts to force urine out into the urethra. The urethra emerges through the penis in males and close to the vagina in females.

Control of urination is caused by the ability to control the sphincter reflex. Up to about 2 years of age children cannot control this reflex and urination is automatic. Once the reflex is controlled, even though the bladder may be very full, urine can be retained for some time.



Normal urine is sterile, but it is easily decomposed by bacterial action outside the body. This results in the formation of ammonia, which is the cause of nappy rash in young children.

Functions of the kidneys

Excretion

The kidneys remove waste products from the bloodstream and convert them to urine. Urine is sent to the bladder for storage and is excreted through the urethra.

Osmoregulation

Water content

The kidneys control the water content of the body. They do this by varying the water content of the urine. For example, on hot days the kidneys conserve water by producing low volumes of urine.

Salt concentration

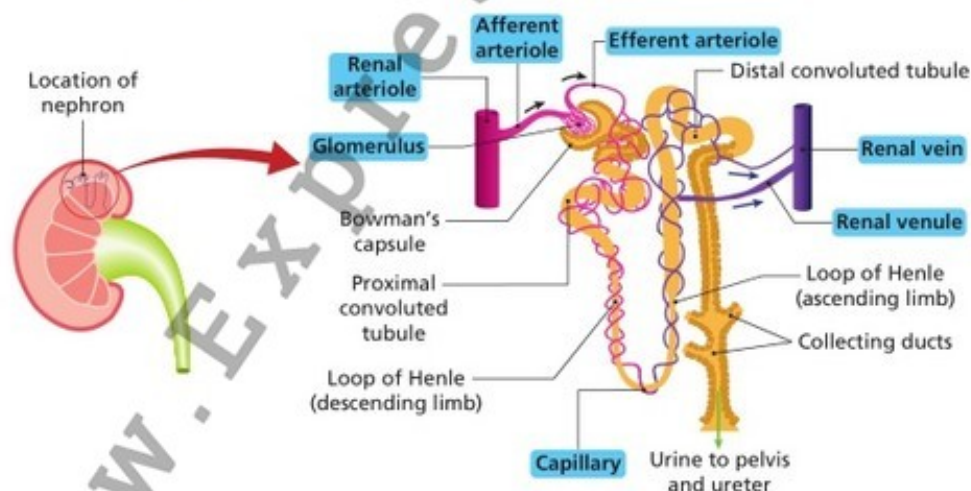
The kidneys control the salt concentration of the body fluids. They achieve this by varying the amount of salt released in the urine. For example, if we consume too much salt the kidneys will increase the amount of salt excreted in urine.

By controlling water and salt concentrations, the kidneys ensure that the blood plasma (and, as a result, all body fluids) has the same concentration as normal body cells. This means that the cells bathed by these fluids do not have problems gaining or losing water by osmosis. For this reason the kidneys are said to regulate osmosis, or to be osmoregulatory.

pH control

The kidneys control the pH of the body fluids. They do this by producing urine that is either more or less acidic. This allows the pH of the blood to remain at its normal value of 7.4.

The nephron



6.6 Structure of the nephron, with its associated blood supply highlighted in blue

Each kidney contains more than a million nephrons. A nephron is a tube about 3 cm long, located in the cortex and medulla of the kidney as shown in diagram 6.6.

Nephrons are the functional units of the kidney, i.e. they make urine. To understand the workings of the kidney it is necessary to understand what happens in each nephron.

Blood supply to the nephron

Blood enters each kidney through the renal artery. Once inside the kidney, this vessel divides to form many renal arterioles, which then split, forming many smaller afferent (incoming) arterioles. Each afferent arteriole in turn divides to form a cluster of capillaries called a glomerulus. A glomerulus is found in each Bowman's capsule, which is a cup-shaped structure at one end of the nephron.

Blood leaves the glomerulus in the efferent (outgoing) arteriole. This then divides to form the capillaries that surround the rest of the nephron. These capillaries eventually re-join to form renal venules, which then combine and emerge from the kidney as the renal vein.

Urine production

Urine is produced in the nephron. There are three main processes involved in urine production: filtration, reabsorption and secretion.

Filtration

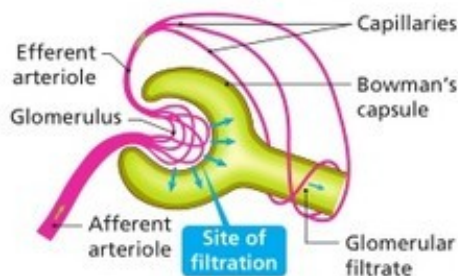
- 1 Blood entering the nephron in the afferent arteriole contains waste products.
- 2 Filtration takes place in the glomerulus. Small molecules such as glucose, amino acids, vitamins, some hormones, urea, salts and water are forced out of the plasma and into Bowman's capsule (diagram 6.9). Here they form a dilute solution called **glomerular filtrate**.
- 3 The structure of the glomerulus helps filtration in three ways:
 - ▶ The pressure in the glomerulus is greater than normal blood pressure. This is caused by the already high pressure of the afferent arteriole being increased due to the efferent arteriole being narrower.
 - Filtration in Bowman's capsule is called **ultra-filtration** because of the high pressure.
 - ▶ The surface area of capillaries in the glomerulus is large. This increases the area for filtration.
 - ▶ The walls of the glomerular capillaries are more porous than normal capillaries. In addition, the wall of Bowman's capsule is only one cell thick.
- 4 Larger substances do **not** enter the glomerular filtrate. These include red and white blood cells, platelets, antibodies, clotting proteins and some hormones.



6.7 Scan of a nephron, showing the glomerulus (red), part of Bowman's capsule (white-brown, at top right) and parts of the tubule (blue)



6.8 Many glomeruli (blue) and blood vessels (red) in a section of the kidney



6.9 Filtration in Bowman's capsule

Filtration means that water and small molecules pass (under high pressure) from the blood into the nephron.

Reabsorption means that molecules pass from the nephron back into the blood.

Active transport means that energy (in the form of ATP) is used to move molecules, often against a concentration gradient, i.e. from low concentrations to high concentrations.

Reabsorption

In the rest of the nephron (beyond Bowman's capsule), water, most salts and useful substances are reabsorbed into the blood. About 99% of the glomerular filtrate is reabsorbed. This leaves about 1.5 litres of urine to be excreted in a normal day.



About 180 litres of glomerular filtrate are formed every 24 hours. This is 4.5 times the fluid content of the body. Obviously not all of this liquid can leave the body as urine.

- In the proximal convoluted tubule:
 - ▶ Most of the water is reabsorbed by osmosis
 - ▶ All of the useful molecules such as glucose, amino acids and vitamins are reabsorbed by a combination of diffusion and active transport
 - ▶ Most of the salts are reabsorbed by active transport or diffusion.

In contrast to active transport, diffusion and osmosis do not require energy. To help in the process of reabsorption the proximal tubule:

 - ▶ Is thin-walled, only one cell thick
 - ▶ Is long (14 mm)
 - ▶ Has numerous infoldings (called microvilli) in its cells
 - ▶ Has a high concentration of mitochondria to provide energy for active transport.
- The descending limb of the loop of Henle is permeable to water. In this section of the loop of Henle a small amount of the water is reabsorbed by osmosis. Also some minerals are reabsorbed.
- The ascending limb of the loop of Henle is permeable to salts. In this region salts move out of the nephron into the fluid of the medulla. Initially the movement of salts is by diffusion, but at the top of the ascending limb sodium is pumped out by active transport. The addition of salts makes the medulla more concentrated than the fluid in the tubule. This helps to remove water (by osmosis) from both the descending limb of the loop of Henle and the collecting ducts. For this reason, the function of the loop of Henle is to reabsorb water.
- The distal convoluted tubule is involved in the delicate, precise control of the water, salt and pH values of the blood. Some water and salts can be reabsorbed from the tubule into the blood in this region.
- The collecting duct is permeable to water. A small amount of water is reabsorbed from the filtrate in the collecting duct. This occurs by osmosis, due to the high salt concentration in the medulla.
- The liquid passing from the collecting duct is called urine. It flows into the pelvis of the kidney and on to the bladder through the ureters.

Secretion

Secretion of potassium ions (K^+) and hydrogen ions (H^+) in the distal tubule help to maintain the pH of blood.

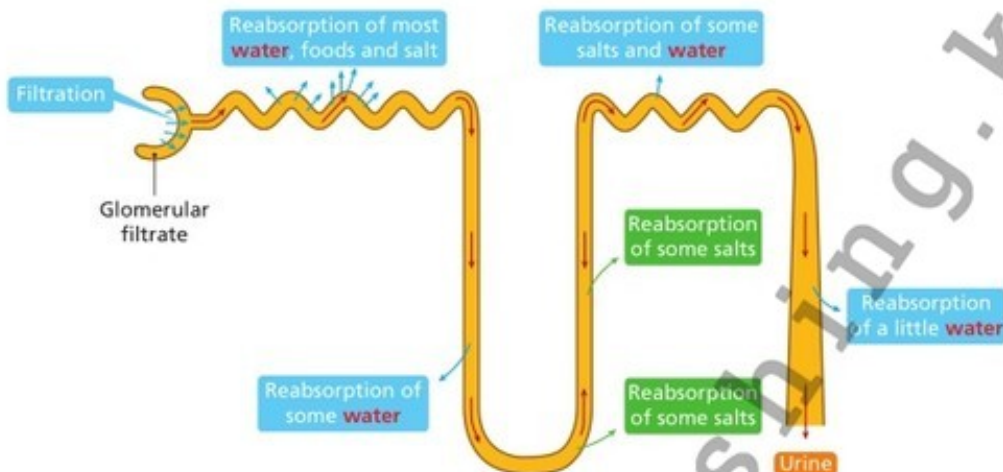


Secretion means that some substances pass from the blood into the nephron.

Summary of nephron functions

The functions of the regions of the nephron, and the materials reabsorbed in each, are summarised in the following table and in diagram 6.10.

Functions of the regions of the nephron		
Region	Substances reabsorbed	Amount of water reabsorbed
Proximal convoluted tubule	Most salts, glucose, amino acids and vitamins	Most
Loop of Henle (descending limb)	Some minerals	A little
Loop of Henle (ascending limb)	Some salts	None
Distal convoluted tubule	Some salts	Some
Collecting duct	None	A little



6.10 Summary of the functions of the nephron

Glomerular filtrate compared with urine

Glomerular filtrate differs from urine in the following ways:

- It has more water (i.e. is more dilute) than urine
- It contains many useful molecules, e.g. glucose and amino acids that are not normally found in urine.

Control of urine volume

ADH

The volume of urine produced is controlled by a hormone called anti-diuretic hormone (ADH, also called vasopressin). ADH:

- Is produced in a part of the brain (the hypothalamus) and stored in the pituitary gland
- Is released from the pituitary gland into the bloodstream
- Affects the distal convoluted tubule and the collecting duct
- Causes more water to be reabsorbed from the nephron
- Controls osmoregulation.

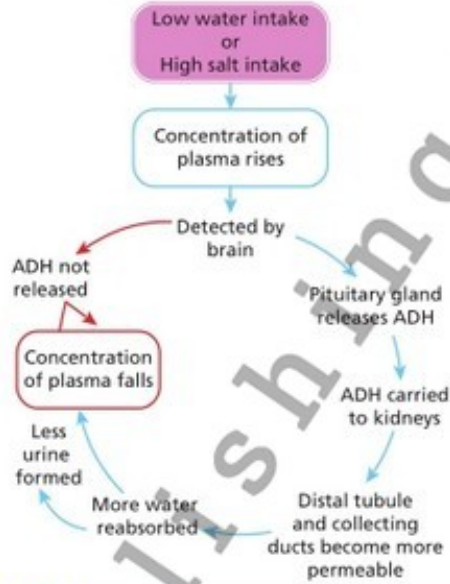
Blood plasma too concentrated

- 1 Blood plasma becomes too concentrated when we:
 - ▶ Drink too little water
 - ▶ Lose too much water as sweat or faeces
 - ▶ Consume too much salt.
- 2 The pituitary gland releases ADH.
- 3 ADH travels to the kidneys in the bloodstream. In the kidneys, ADH causes the walls of the distal tubule and the collecting ducts to become more permeable to water.
- 4 More water is reabsorbed from the nephron. This causes a reduction in:
 - ▶ The salt concentration of the plasma
 - ▶ The volume of urine produced.

Blood plasma concentration normal or too dilute

- 1 Blood plasma concentration is normal or too dilute when we:
 - ▶ Consume a great deal of water
 - ▶ Eat a low-salt diet.
- 2 In this situation ADH is not released.
- 3 The distal tubules and collecting ducts become relatively impermeable to water.
- 4 Very little water is reabsorbed from the distal tubules and collecting ducts. This causes:
 - ▶ The concentration of the plasma to remain relatively unchanged
 - ▶ The production of a large volume of urine.

The effects of different conditions on urine production are shown in the table below.



6.11 How ADH controls urine volume

Effects of different conditions on urine production				
Condition	Effect on blood	ADH	Distal tubule and collecting duct	Urine
Thirsty or Salty diet or Hot day or Exercise or Sweating	Low water content and high salt concentration	Released	More permeable to water	Low volume of water; higher salt concentration (i.e. a low volume of concentrated urine)
Excessive water intake or Very low-salt diet	High water content and low salt concentration	Not released	Less permeable to water	High volume of water; lower salt concentration (i.e. a high volume of dilute urine)
High protein diet	Normal water content and increased concentration of urea	No effect	No effect	Same volume of water; increased urea concentration (i.e. the same volume of concentrated urine)

Dialysis

When kidneys fail, waste products cannot be removed adequately from the body and start to build up. An increased level of waste products in the blood is a condition called 'azotemia,' and as waste products build up they cause people to experience a sick feeling throughout the body which is called 'uremia.'

Dialysis is a procedure that is a substitute for many of the normal functions of the kidneys. It allows patients to lead relatively normal lives, even though their kidneys are no longer working adequately. Doctors can use a variety of tests to establish whether a person is in need of dialysis. One of the main ones involves measuring the level of 'creatinine' in both the blood and the urine. A comparison of the two measures indicates the extent to which the kidneys are failing to excrete waste products from the body.

In cases where the 'creatinine clearance' measure indicates that someone is in need of dialysis, there are two different types of dialysis a person can follow: hemodialysis or peritoneal dialysis.

Hemodialysis

In this form of treatment, blood is passed out of the body and through a filter in a dialysis machine. A tube is placed between an artery and vein in the patient's arm or leg and waste products are filtered out in the machine and then the blood passes back into the body.



6.12 Hemodialysis procedure

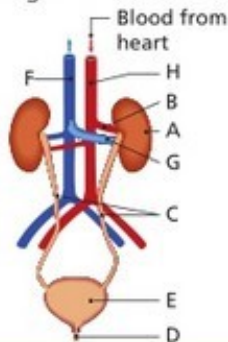
Peritoneal dialysis

This form of dialysis focuses on a membrane which lines the abdominal cavity: the peritoneal membrane. A plastic tube is inserted through the abdominal wall and into the abdominal cavity. A special fluid is then flushed into the abdominal cavity and then washes around the intestines. The peritoneal membrane then acts a filter between this fluid and the bloodstream, removing excess water and waste products in the process.

Research using the internet either (a) the advantages and disadvantages of each type of dialysis described above or (b) the advantages and disadvantages of kidney transplant compared to treatment by dialysis.

Questions on Module 6

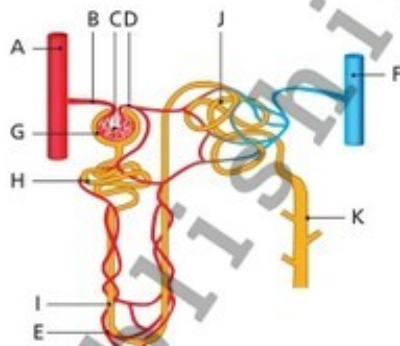
- What is meant by:
 - Excretion
 - Metabolism
 - Homeostasis?
- Name three human excretory organs.
 - State the main substances excreted by each organ.
 - Why is faeces not considered to be excreted?
- Name the parts labelled A to I in diagram 6.13.



6.13 The urinary system

- State the part in the diagram that carries out each of the following:
 - Makes urine
 - Stores urine
 - Carries blood to the kidney
 - Takes blood from the kidney
 - Controls the release of urine
 - Excretes urine.
- Why are the kidneys referred to as osmoregulatory structures?
 - Name two ways by which the body gains water.
 - Name four ways by which water is lost from the body.
 - From what food type is urea made?
 - In what organ is urea made?
 - Where does urea enter the blood?
 - Where does urea leave the blood?
 - Where does urea leave the body?
 - Where in the body is urine
 - Made
 - Stored
 - Excreted?
 - Draw a labelled diagram to show the location of a nephron in the kidney.

- Redraw diagram 6.14 twice the size shown here.
- On your diagram name the parts A to K labelled on diagram 6.14.
- Give one function carried out by each part labelled.



6.14 The nephron

- Why is the blood pressure in the glomerulus higher than normal?
 - Why do the cells lining some parts of the nephron contain many mitochondria?
- The table below shows the composition of plasma, glomerular filtrate and urine.

Substance	% in plasma	% in glomerular filtrate	% in urine
Protein	8	0	0
Glucose	0.2	0.2	0
Urea	0.03	0.03	2
Salts	0.7	0.7	1.5

- Why is there no protein in the glomerular filtrate or urine?
- Explain why the urine has no glucose.
- According to these figures, the urea concentration in the urine is much greater than that in the glomerular filtrate. Explain why this is so.
- Why does the salt concentration only increase by a factor of about 2 (i.e. 0.7% to 1.5%) while urea concentration increases by a factor of 67 (i.e. 0.03% to 2%)?

- 10 Describe the effects on the composition and volume of urine of the following:
- A hot day
 - Drinking a lot of water quickly
 - Eating a protein-rich meal.
- 11
- Name a hormone that controls urine volume.
 - From which gland is this hormone released?
 - State two situations that might cause it to be released.
 - Name the parts of the nephron affected by the hormone.
 - What effect has a high level of the hormone on (i) Urine (ii) Blood plasma?
- 12 The functions of the nephron can be summarised as filtration, reabsorption and secretion.
- Explain the meaning of each of the underlined terms.
 - State a location and name two substances involved in filtration and reabsorption.
- 13 Dialysis is procedure that can replace some of functions of failing kidneys.
- Which chemical in blood and urine is screened to indicate a patient's need for dialysis treatment?
 - Name two forms of dialysis.
 - Identify one advantage of each form of dialysis.
- 14 Suggest why we produce a larger volume of urine when we exercise vigorously, even when we do not drink any more water than usual.
- 15 The filtrate in the kidney tubule finally becomes urine in the collecting duct. List, in order, the structures that urine passes through from the collecting duct to leaving the body.
- 16 Many hormones do not pass into urine in the kidneys. Some pregnancy tests detect the levels of hormones in urine. What does that suggest about the hormones used in pregnancy testing compared with the ones that do not end up in urine?
- 17 Suggest why consuming too much salt makes us feel thirsty.
- 18 State and explain the effect on volume and concentration of urine produced when drinking excess water.

Module 7 The cell cycle

Learning objectives

- Investigate the phases of mitosis and examine mitotic activity under a microscope (10.2.2.1)
- Explain features of gamete formation in plants and animals (10.2.2.2)
- Explain the occurrence of tumours by uncontrolled cell division (10.2.2.3)
- Explain links between mitosis and the ageing process (10.2.2.4)

Cell continuity



Cell continuity means that all cells develop from pre-existing cells.

Cell continuity gives rise to the continuity of life.

When a new cell forms (from an existing cell) it goes through the following three phases:

- It produces or synthesises all the materials it will need
- It grows larger
- It reproduces to form new cells.

Cell continuity implies that most cells spend a lot of time producing the chemicals and substances they need to survive and grow. They are not actively dividing into new cells during this phase of their life. Cells spend a relatively short time engaged in cell division.

Chromosomes

When a cell is not dividing, the chromosomes exist as long, thin threads called chromatin. At cell division, chromatin contracts to form a number of clearly distinguishable chromosomes.

Every species has a definite number of chromosomes in each cell. For example, humans have 46 chromosomes in each body cell.

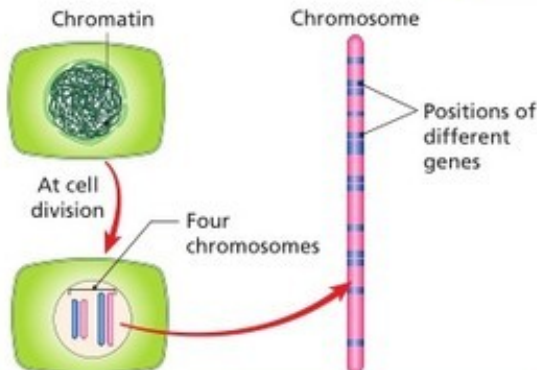
Each chromosome is composed of hundreds, or even thousands, of genes. These genes are arranged along the chromosome as shown in diagram 7.2.



Chromosomes are coiled threads of DNA (which forms genes) and protein that become visible in the nucleus at cell division.



7.1 A human chromosome (SEM)



7.2 Chromatin, chromosomes and genes

Genes

Many of the proteins produced by genes are enzymes. As these enzymes control the activities of the cell, it can be said that genes (or chromosomes) control the cell. Genes are also said to be units of inheritance.

All the genes in an organism make up its **genome**. In humans, genes control features such as eye colour, production of skin pigment (melanin), number of fingers, the shape of the face and about 20 000 to 25 000 other features. In plants, genes control petal colour, leaf shape, fruit taste and many more features.

A **gene** is a section of DNA that contains the instructions for the formation of a protein.

Haploid and diploid cells

A **haploid cell** has one set of chromosomes, i.e. it has only one of each type of chromosome in the nucleus.

A **diploid cell** has two sets of chromosomes, i.e. it has two of each type of chromosome in the nucleus.

Haploid is symbolised by the letter 'n', and the number of chromosomes in the cell is given as $n = 2$ or $n = 3$ etc. In diagram 7.3, all the cells shown are haploid because in each case there is only one of each type of chromosome.

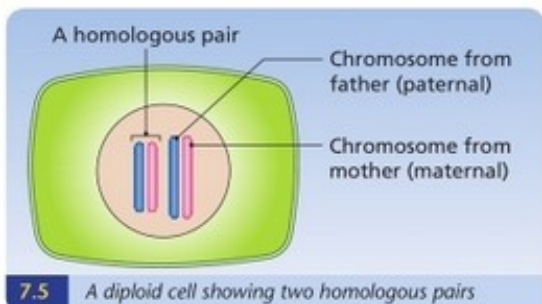
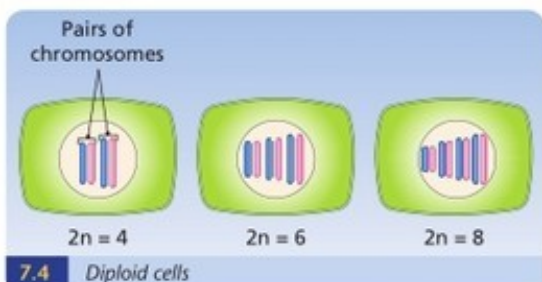
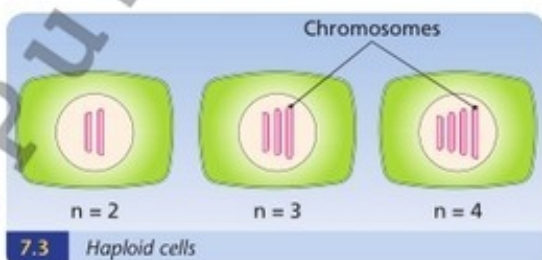
In humans, eggs and sperm are haploid cells and each one contains 23 chromosomes (i.e. $n = 23$).

Diploid is symbolised as '2n', and the total number of chromosomes in the cell is given as $2n = 4$ or $2n = 6$, etc.

The diploid number for humans is 46 ($2n = 46$). This means that each human cell has 23 chromosomes that were obtained from the person's mother and 23 that were obtained from the father.

In a diploid cell the chromosomes are in pairs. As each pair of chromosomes has similar genes, they are called **homologous pairs**.

In diploid cells, one chromosome from each homologous pair is derived from the mother and the other one from the father. This is shown in diagram 7.5, in which the maternal and paternal chromosomes are shown in different colours.

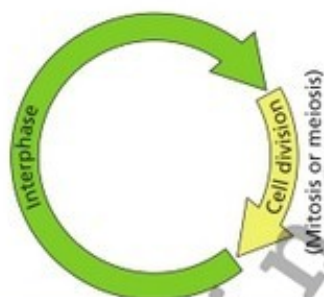


A **homologous pair** is two chromosomes of similar size with the same sequence of genes.

The cell cycle

A small number of cells, such as nerve and red blood cells, do not divide when they reach full size. Most cells, however, grow until they reach a certain size and then divide.

The cell cycle describes the life cycle of a cell. At its simplest, the cell cycle is divided into a period when the cell is not dividing, called interphase, and a period when the cell divides, called mitosis (or meiosis).



7.6 The cell cycle

The cell cycle is the changes that take place in a cell during the period between one cell division and the next.

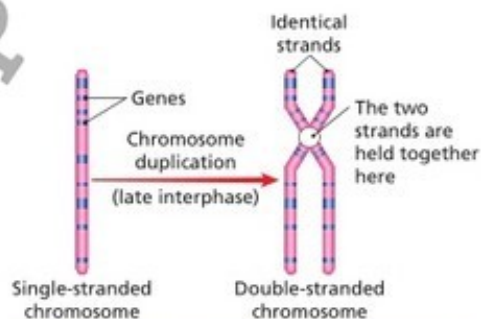
Interphase is the phase in the cell cycle when the cell is not dividing.

Interphase is the longest phase in the cell cycle, often accounting for over 90% of the cycle.

During interphase, the chromosomes are very elongated. It is not possible to distinguish individual chromosomes in the nucleus during interphase. Instead, they appear as a mass of material called chromatin, as shown in diagram 7.2.

Although cells are not dividing during interphase, they are very active in the following ways during this phase.

- In the early part of interphase the cell is very active, producing new organelles such as mitochondria or chloroplasts. It also forms many chemicals that are needed for growth, especially enzymes and other proteins.
- Towards the end of interphase (just before the cell divides), the chromosomes produce identical copies of themselves. The duplication (or doubling) of a chromosome produces a chromosome with two strands. The two strands have identical genes, as shown in diagram 7.7.



7.7 Chromosome duplication

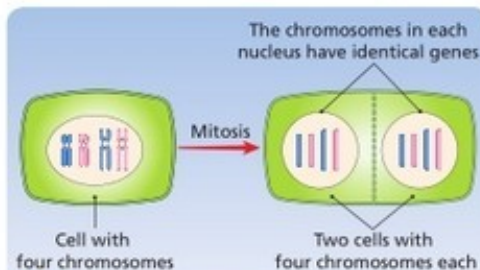
Mitosis

Each of the new nuclei formed in mitosis becomes enclosed within a cell called a daughter cell, even when the process occurs in male cells.

Mitosis is a form of nuclear division in which one nucleus divides to form two nuclei, each containing the same number of chromosomes with identical genes.

The two daughter cells each have the same number of chromosomes. Not only do they have the same number of chromosomes, but the genes on the chromosomes in each cell are identical to each other.

Mitosis takes place in cells that are not associated with the reproductive system. These cells are called somatic cells.



7.8 Summary of mitosis

Mitosis produces two cells that are identical to each other in terms of chromosome numbers and the genes present on the chromosomes.

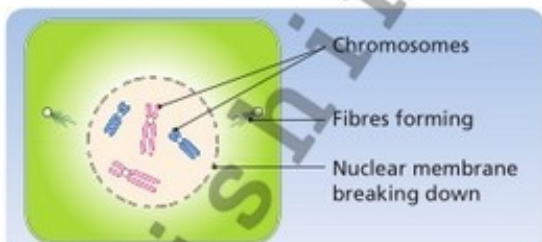
Stages of mitosis

Mitosis is a continuous process that is often described as if it had four definite stages or phases. Each stage runs smoothly into the next and it is often difficult to say exactly when any stage starts and ends.

The account of mitosis that follows refers to an animal cell with four chromosomes, i.e. an animal cell with a diploid number of four ($2n = 4$).

Stage 1

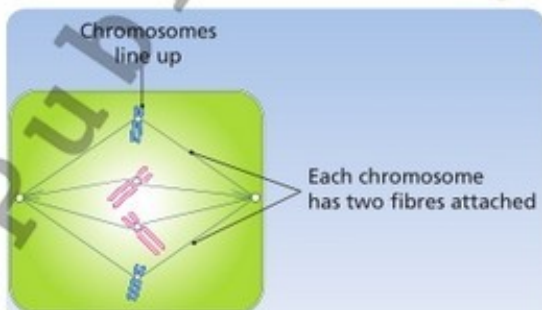
- At the end of interphase, and early in stage 1, chromosomes contract. They gradually become visible in the nucleus as short, thickened strands.
- Each chromosome appears as a double strand.
- Fibres begin to appear in the cytoplasm of the cell.
- The nuclear membrane starts to break down.



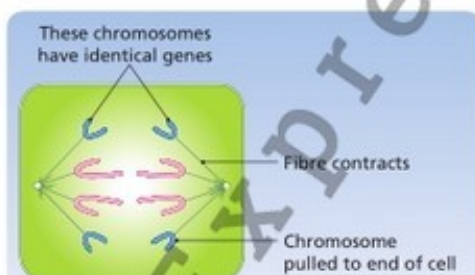
7.9 Stage 1

Stage 2

- The nuclear membrane is fully broken down.
- The chromosomes move so that they line up across the middle of the cell.
- Two fibres attach to each of the chromosomes.



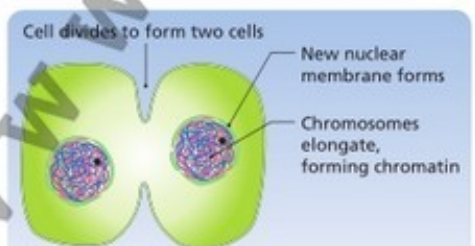
7.10 Stage 2



7.11 Stage 3

Stage 3

- The fibres contract. This means that each chromosome is pulled apart.
- The two strands within each chromosome are pulled to opposite ends of the cell.
- Each of the strands in a double-stranded chromosome has identical genes. As a result of the chromosomes splitting at this stage, an identical set of genes is pulled to each end of the cell.



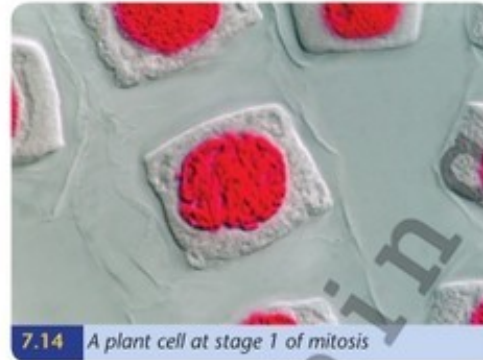
7.12 Stage 4

Stage 4

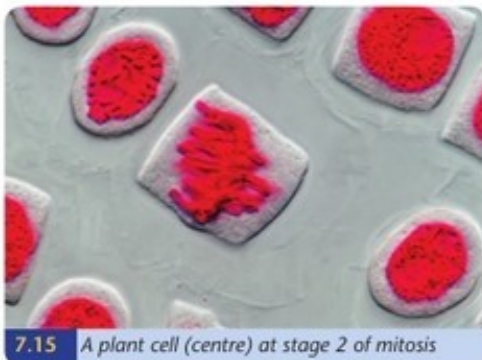
- A nuclear membrane forms around each of the two sets of chromosomes.
- The chromosomes elongate within each nucleus and become chromatin.



7.13 Plant cells in interphase



7.14 A plant cell at stage 1 of mitosis



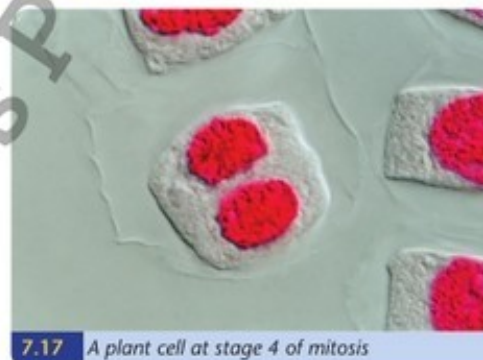
7.15 A plant cell (centre) at stage 2 of mitosis



7.16 A plant cell (centre) at stage 3 of mitosis

Cell division

Once mitosis is complete, the original cell divides to form two cells. These daughter cells will contain the same number of identical chromosomes as each other. In addition, each daughter cell will have some (about half) of the cell organelles and biomolecules that were in the parent cell.



7.17 A plant cell at stage 4 of mitosis

Function of mitosis

Unicellular organisms

In single-celled (unicellular) organisms mitosis increases the number of individuals and is used as a **method of reproduction**. This is the case in organisms such as **Amoeba** and bacteria.

Reproduction that does not involve the joining of two cells is called asexual reproduction. In other words, asexual reproduction only involves a single parent or cell. Mitosis is the basis of asexual reproduction.

Multicellular organisms

In many-celled (multicellular) organisms mitosis is responsible for **growth and repair of cells**. In these organisms mitosis produces new cells, not new individuals.

Mitosis is responsible for the single-celled zygote growing into an embryo.

Even when a person is fully grown, mitosis is essential to replace old and damaged cells. This is seen when new blood cells are produced, when skin damaged by a cut is repaired or when torn muscles are healed.

Mitosis is also responsible for growth and repair in plants.

Demonstration: To observe mitosis in an onion root tip**Materials**

- | | |
|-------------------------|--------------------------------------|
| 1 Onion plant with root | 5 Pasture pipette |
| 2 Acetocarmine stain | 6 Alcohol lamp |
| 3 Scissors | 7 Microscopic slides and cover slips |
| 4 Forceps | 8 Light Microscope |

Procedure

- 1 Take the onion plant with newly sprouted roots.
- 2 Cut from the tip of a root a length of about 5 to 8 mm.
- 3 Place this on a clean microscope slide and add 2-3 drops of acetocarmine stain to the slide.
- 4 Warm the slide gently over the alcohol lamp for about one minute. The slide should only be warmed to the touch and the root should not be allowed to dry out.
- 5 Use a cover slip to cover the slide.
- 6 Flatten the root in the slide but pressing down firmly but so not press so hard that the cover slip slips or breaks.
- 7 Place the slide under a compound microscope in 10x objective. Scan and narrow down to a region containing dividing cells and use 40x to observe more closely.



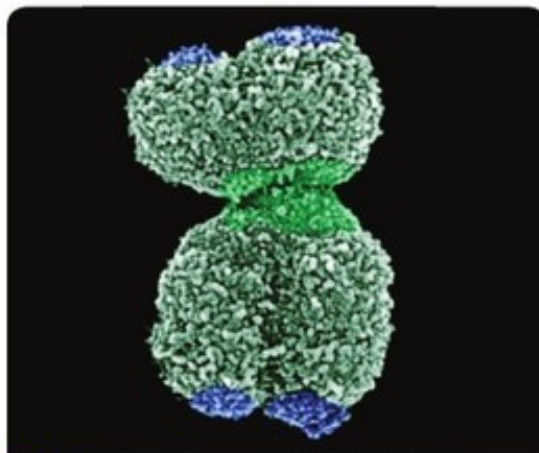
7.18 A light microscope

Detailed study of mitosis

The account of mitosis that follows is for a similar cell to that outlined earlier in this module (i.e. an animal cell with four chromosomes).

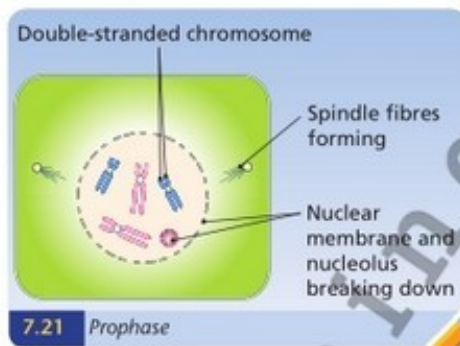
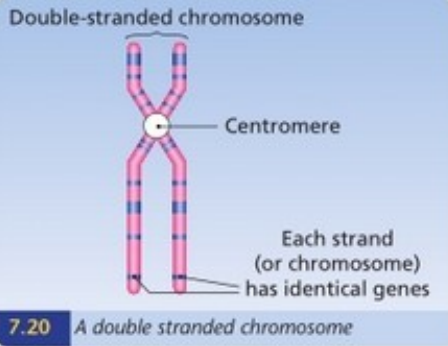
Prophase

- At the end of interphase and early in prophase chromatin starts to contract.
- Chromosomes become visible as double-stranded structures. The point at which the strands are held together is called a **centromere**.
The two strands in a chromosome have identical genes. In fact, each strand is a chromosome.
- The nucleolus disappears. The nucleolus is a region in the nucleus where ribosomes are made. Some cells have more than one nucleolus.
- The fibres that appear in the cytoplasm at this stage are called spindle fibres. All the spindle fibres collectively form a structure called the spindle.
- The nuclear membrane starts to break down.



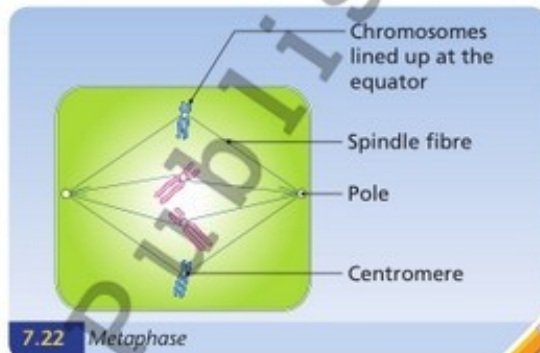
7.19 A human chromosome (SEM) showing the centromere (bright green)

The centromere is the point at which the chromosomes are attached in a double-stranded chromosome.



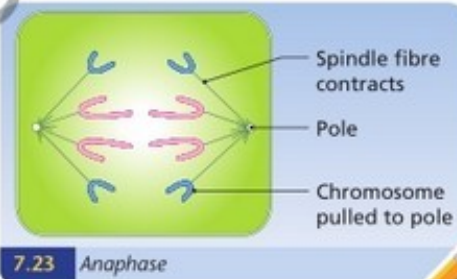
Metaphase

- In metaphase the nuclear membrane completes its breakdown.
- A spindle fibre from each end (or pole) of the cell attaches to each centromere.
- The chromosomes line up across the middle, or equator, of the cell.



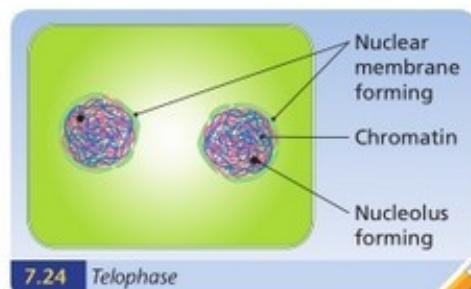
Anaphase

- The spindle fibres contract. This causes the centromeres to split.
- One strand (or chromosome) from each double-stranded chromosome is pulled to opposite poles of the cell. This means the cell has eight chromosomes at this stage. The four chromosomes pulled to each pole have identical genes.
- Anaphase is the shortest phase in mitosis. It often lasts only a few minutes, compared with up to 30 minutes for each of the other phases.



Telophase

- The four chromosomes at each pole begin to lengthen and become hard to distinguish.
- The spindle fibres break down.
- One or more nucleoli (singular: nucleolus) begin to re-form.
- A nuclear membrane forms around the chromatin at each end of the cell.
- At the end of mitosis the original nucleus has divided into two identical nuclei.



Cancer



Cancer is a group of disorders in which certain cells lose their ability to control both the rate of mitosis and the number of times mitosis takes place.

Normally the rate of mitosis and cell division is carefully controlled. This means that just enough new cells are formed to allow for normal growth and repair.

However, sometimes a cell (or a group of cells) loses the ability to control the rate of mitosis. A mass of cells forms, called a tumour. Tumours may be benign or malignant.

Benign tumours

Benign means 'kind'. In a benign tumour the cells stop dividing after some time. Benign tumours are not life-threatening. They do not invade other tissues.

Examples of benign tumours are warts (caused by a virus) and skin 'tags' (small blobs of raised skin). Most breast tumours are benign. Benign tumours can be surgically removed.

Malignant tumours

Cancer results in an uncontrolled multiplication of abnormal cells. These abnormal cells form a malignant tumour.

Malignant tumours, called 'cancers', may be life-threatening. This is because they can invade other cells and can move from one place to another in the body. This movement (or migration) of malignant cells is called metastasis.

Cancer cells continue to divide indefinitely. For this reason they are said to be 'immortal'.

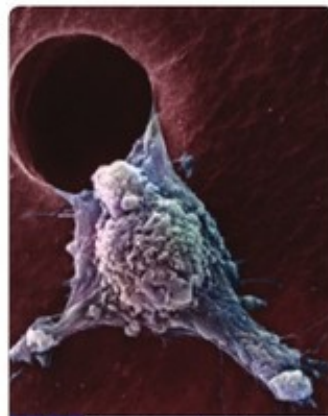


7.26 An examination for skin cancer

Causes of cancer

Cancer is caused when normal genes are altered to form cancer-causing genes (called **oncogenes**). These alterations are brought about by cancer-causing agents called carcinogens. Some common **carcinogens** are cigarette smoke, asbestos fibres, dioxins, pesticides, ultraviolet radiation and some viruses.

It is important to realise that most cancers can be cured. This is especially so if they are discovered and treated early. Treatment includes surgery, radiation (to burn out the cancer) and the use of chemicals that slow down mitosis (chemotherapy).



7.25 A cancer cell on the move

Meiosis



Meiosis is a form of nuclear division in which the four daughter nuclei contain half the chromosome number of the parent nucleus.

Immediately after meiosis, the daughter nuclei are enclosed by cells.

When meiosis takes place in a diploid cell, all daughter cells will be haploid. If these cells are capable of joining with another haploid cell from the opposite sex, they are called sex cells or **gametes**.

Most human cells have 46 chromosomes. Meiosis occurs in the ovaries and testes to produce gametes called eggs and sperm, respectively. As a result of meiosis, there are 23 chromosomes in each egg or sperm.

Functions of meiosis

Meiosis has two basic functions in multicellular organisms:

- It allows for sexual reproduction, involving gametes, while still maintaining the parental chromosome number
- It allows for new combinations of genes to be formed, which will give rise to variations among organisms.

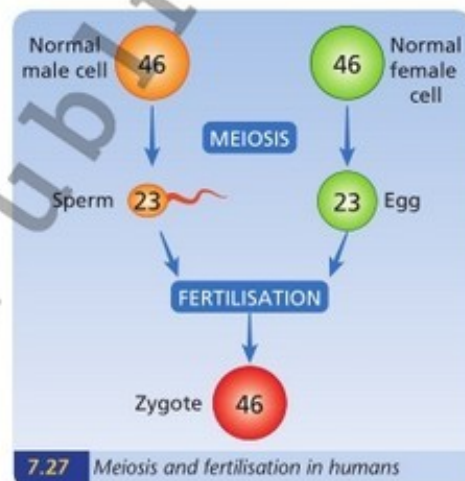
Meiosis allows for sexual reproduction

In sexual reproduction, two haploid (n) cells join to form a diploid ($2n$) zygote. These haploid cells are called gametes or sex cells.

In animals, the gametes are the sperm and egg. In flowering plants, the gametes are the male gamete nuclei and the egg and polar nuclei. The zygote will contain the normal number of chromosomes.

Meiosis is essential for sexual reproduction because it halves the chromosome number. This means that the normal chromosome number is restored at fertilisation.

The role of meiosis in humans is outlined in diagram 7.27. The normal human chromosome number per cell is 46. Meiosis halves this number to 23 in the gametes. Fertilisation restores the chromosome number to 46.



7.27 Meiosis and fertilisation in humans

Meiosis allows for variation

The cells resulting from meiosis are not identical. Their genes vary due to the exchange of genetic material which takes place during meiosis.

The variation in the genes (genetic variation) produced in meiosis results in variations or differences in the organisms resulting from the re-assortment of genetic material due to sexual reproduction. This is why brothers (or sisters), while they may resemble each other, are rarely identical.

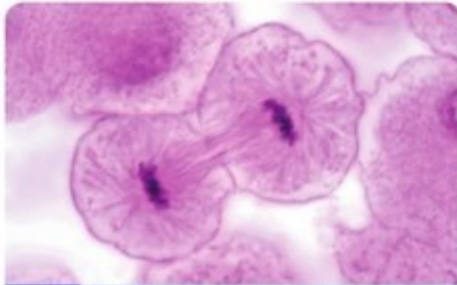
Variations produced in this manner are part of the basis of evolution, as detailed in Module 8.

The differences between mitosis and meiosis

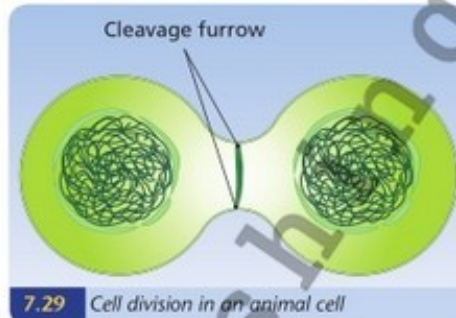
Mitosis	Meiosis
The daughter cells have the same number of chromosomes as the parents	The daughter cells have half the number of chromosomes of the parents
The daughter cells have identical genes on their chromosomes	The daughter cells have different genes on their chromosomes
Two cells are formed	Four cells are formed

Cell division

Cell division follows immediately after mitosis. The process of cell division proceeds differently in animal and plant cells.



7.28 A cell showing a cleavage furrow



7.29 Cell division in an animal cell

Cell division in animal cells

Cell division occurs in animals by a process called cleavage. A shallow groove, called a **cleavage furrow**, appears around the cell, lining up with the position occupied by the equator during metaphase.

The cleavage furrow becomes deeper, until it eventually divides the cytoplasm and the cell splits into two.



Animal cell →
cleavage furrow forms
Plant cell → cell
plate forms

Cell division in plant cells

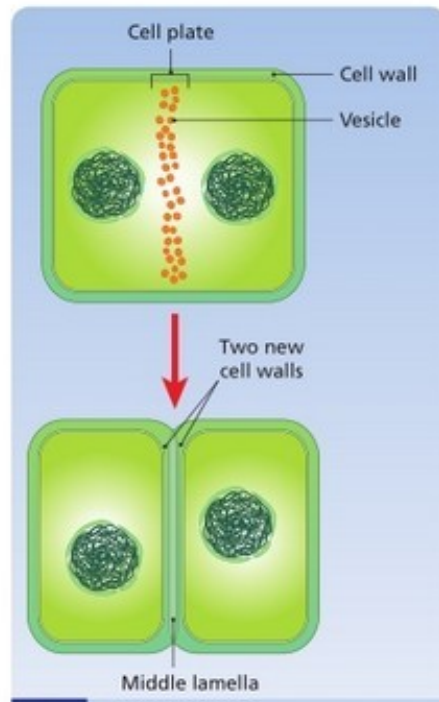
In plant cells, the rigid cell wall prevents a cleavage furrow from forming. Instead, a number of small membrane-enclosed sacs, called vesicles, gather in the area between the two nuclei. The vesicles contain the material, mainly cellulose, which forms the new cell walls, as shown in diagram 7.31.

These vesicles form a structure called the **cell plate**. The cell plate enlarges and its membranes join with the cell membrane of the original cell.

Two cell walls form from the cell plate, one for each of the daughter cells. The region between two adjacent plant cell walls is called the **middle lamella**.



7.30 A cell plate in a dividing onion cell



7.31 Cell division in a plant cell



Research the link between mitosis and ageing. What happens in the cell cycle process that leads to ageing?

Gametogenesis



Read the text and review the diagrams below. Then complete the table which summarises the similarities and differences between the gametogenesis processes of males [spermatogenesis] and females [oogenesis]

	Spermatogenesis	Oogenesis
Gametes		
Location of the process		
Meiotic divisions		
Number of gametes produced		
Duration of process		
Beginning of process		
End of process		

Gametogenesis is the process by which diploid precursor cells through meiotic division become haploid gametes (sex cells).

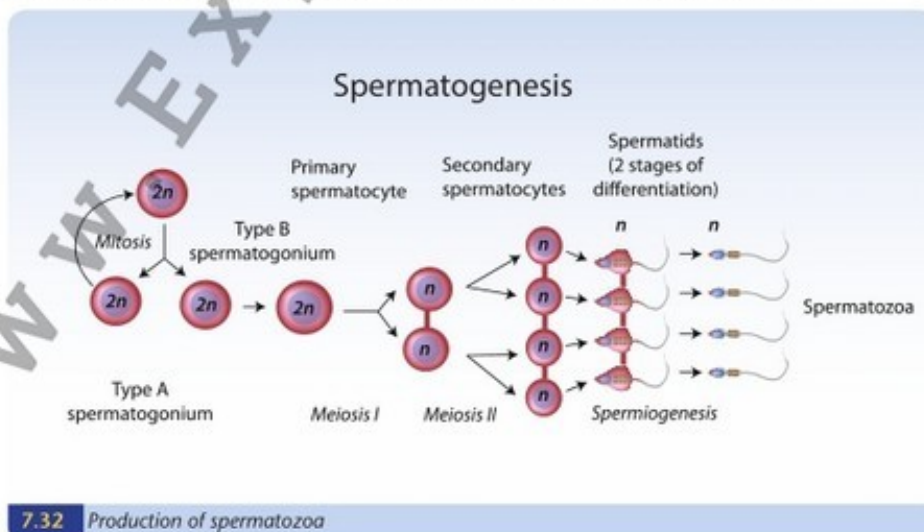
The process in males, which is called *spermatogenesis*, produces spermatozoa (sperm). In females, the process called *oogenesis*, produces ova (eggs).

Gametogenesis occurs in the gonads. The process involves these phases:

- Multiple mitotic divisions and cell growth of precursor germ cells
- Meiotic divisions (I and II) to produce haploid daughter cells
- Haploid daughter cells which undergo differentiation to become functional gametes

Spermatogenesis

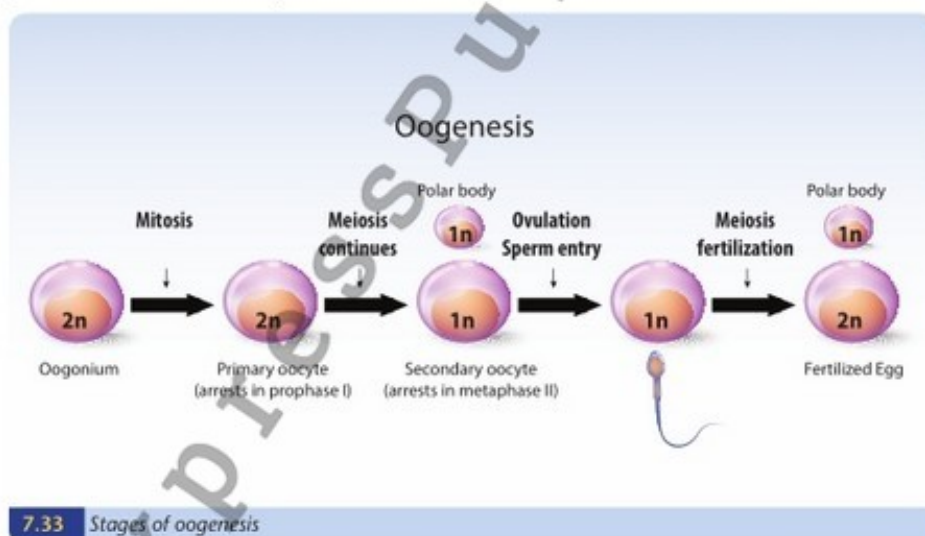
- The production of spermatozoa (sperm) takes place in the testes.
- The process begins at puberty when diploid spermatogonia go through mitosis and a period of cell growth in developing into spermatocytes.
- The spermatocytes then go through two meiotic divisions to form four haploid daughter cells (*spermatids*).
- This is followed by a process of differentiation through which the spermatids develop into functional sperm cells (*spermatozoa*).
- This process is continuous throughout the life of the male but slows with age.



7.32 Production of spermatozoa

Oogenesis

- The production of female gametes (ova) takes place partly in the oviduct but mainly within the **ovaries**.
- During foetal development, a large number of primordial cells are formed by mitosis.
- These cells (*oogonia*) go through cell growth and then through meiosis become *primary oocytes*.
- This development of the cell is arrested in the first prophase stage and from birth until puberty the cell remains in prophase 1.
- At the point at which the female begins the menstrual cycle, hormones trigger the continued division of some of the primary oocytes each month.
- The cell divides unequally, with most of the cellular material and organelles going to one cell, called a secondary oocyte. The second cell forms a polar body that usually dies.
- The secondary oocyte starts the second meiotic division which is again arrested in metaphase II.
- At ovulation, this secondary oocyte will be released and moves through the oviduct toward the uterus.
- If the secondary oocyte is fertilized by a sperm, meiosis II will be completed. A second polar body and a fertilized egg which contains all 46 chromosomes of a human being are produced.
- The process ends with menopause in the female.

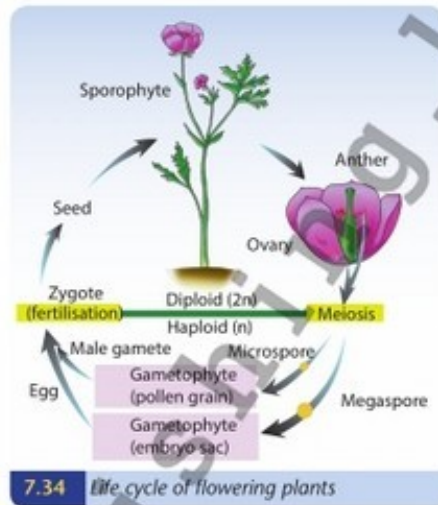


The life cycle of flowering plants

Flowering plants have two main stages in their life cycles: the **sporophyte** and the **gametophyte** stages. In the sporophyte stage of the flowering plant, the plant has a full set of chromosomes and is referred to as being diploid. The plant has developed from a zygote (fertilised egg). In flowering plants, the sporophyte stage is the adult plant with a root and shoot system. The sporophyte produces spores by meiosis (a type of cell division that reduces the chromosome number by half). These spores develop into the gametophyte stage in the flower. Flowers have both male and female parts. The male part (stamen) produces the male gamete and the female part (carpel) contains the ovaries that produce the female gametes. The male and female gametes have half the number of chromosomes of the parent plant. The gametes are referred to as being haploid. This is the second stage of the plant's life cycle (gametophyte) and it is much smaller than the sporophyte stage.

On fertilisation, the male gamete fuses with the female gamete to produce a diploid zygote and restores the chromosome number to being diploid. It now enters the sporophyte stage of its life cycle, where the plant has a full set of chromosomes. The zygote grows and develops into a seed. Once the seed germinates it will grow into a plant. As a result the plant spends only a brief amount of time as a gametophyte and the remainder of its life is spent as a sporophyte.

Sporophyte is the diploid stage in the life cycle of a plant. The sporophyte produces spores by meiosis. **Gametophyte** is the haploid stage in the life cycle of a plant. The gametophyte stage produces the male gametes (in pollen) and the female gametes (egg cells in the ovaries of the flower).



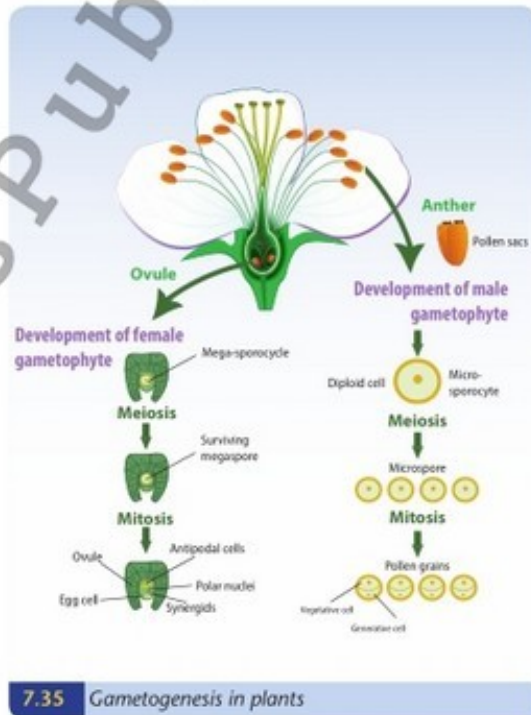
7.34 Life cycle of flowering plants

Gametogenesis in plants

Plant life cycles alternate between sporophyte and gametophyte generations. Meiosis produces spores which then undergo gametogenesis.

The diagram shows that the male gametophyte develops within the anthers of a flower. It is the pollen grain plus the pollen tube that grows from it. The pollen tube delivers haploid nuclei (sperm) to a haploid nucleus (egg) within the embryo sac of the flower. Four microspores are produced by meiosis and these microspores then separate and develop into pollen grains. The generative nucleus then divides by mitosis to form two haploid nuclei, [sperm nuclei].

The female gametophyte develops within an ovule contained in the ovary of the pistil. Within the ovule, meiosis produces haploid cells. One of these cells grows to become a megaspore. Its nucleus will divide by mitosis to produce a multinucleate structure known as the embryo sac. This structure develops into the mature embryo [the female gametophyte].



7.35 Gametogenesis in plants

Questions on Module 7

- Explain what is meant by:
 - Cell continuity
 - The continuity of life
 - The cell cycle.
- With regard to chromosomes:
 - What substances are they made of?
 - Where in the cell are they located?
 - What is their function?
 - How many chromosomes are present in a normal human cell?
- What is a gene?
 - Name the substance of which genes are made.
 - Use a diagram to explain the relationship between genes and chromosomes.
 - State the function of genes.
- Draw simple diagrams to show each of the following cells:
 - The chromosomes are single-stranded and the cell has a diploid number of six.
 - The chromosomes are double-stranded for a cell where $n = 3$.
 - The chromosomes are double-stranded for a cell where $2n = 4$.
- Name the main periods in the cell cycle.
 - Explain why interphase is not a resting phase in the cell cycle.
- Refer to diagram 7.36 and answer the following:

7.36 Stage of mitosis

 - How many centromeres are shown in the diagram?
 - Is the cell shown haploid? Give a reason for your answer.
- Name two events that should occur immediately before the stage of mitosis shown.
 - Draw a labelled diagram of the cell immediately after the stage of mitosis shown.
 - When this cell has finished mitosis, how many chromosomes will be in each nucleus?
- Distinguish between interphase and mitosis under the headings:
 - Duration
 - Chromosome appearance
 - What happens to organelles and chromosomes.
- With regard to mitosis, state:
 - Why the nuclear membrane must break down
 - Why fibres form
 - How the chromosomes are pulled apart
 - How many chromosomes will be in each resulting nucleus.
- Are the cells in a person's body all identical genetically?
- A cell has a chromosome diploid number of 4. Draw a labelled diagram to show this cell in the second stage of mitosis.
- What is a tumour?
 - Distinguish between benign and malignant tumours.
 - What is cancer?
 - Give two reasons why cancer cells are said to be abnormal cells.
- What are carcinogens?
 - Name two common carcinogens.
- A cell has 46 chromosomes. How many chromosomes will be in the nuclei as a result of:
 - Mitosis
 - Cancer
 - Meiosis?
- If meiosis did not occur, why would sexual reproduction be a problem?

- 15 A chimpanzee has 48 chromosomes per cell. How many chromosomes are in each of the following:
- Chimpanzee eggs
 - Chimpanzee sperm
 - Chimpanzee zygote
 - Chimpanzee mouth cell?
- 16 Name the stages in mitosis associated with the following events:
- The formation of spindle fibres
 - Nuclear membrane formation
 - Contraction of spindle fibres
 - Lining up of chromosomes
 - Movement of chromosomes to the poles
- 17 Draw a labelled diagram of a plant cell at metaphase of mitosis. Show the chromosome number as $2n = 6$.
- 18 State which of the options that follow, (i), (ii), (iii) or (iv), is the correct answer.
- In human liver cells, the correct chromosome number is:
(i) 23 (ii) 24 (iii) 92 (iv) 46.
 - Genes are made from:
(i) Chromosomes (ii) DNA
(iii) Chromatin (iv) Proteins.
 - The cell cycle consists of:
(i) Four stages (ii) Three stages
(iii) Two stages (iv) Thousands of stages.
 - Chromatin consists of:
(i) Cytoplasm (ii) Centromeres
(iii) DNA and protein
(iv) Spindle fibres.
- A cleavage furrow develops after:
(i) Prophase (ii) Metaphase
(iii) Anaphase (iv) Telophase.
 - A cell plate develops in:
(i) Metaphase (ii) Plant cells
(iii) Cancer cells (iv) Animal cells.
- 19 "All the genes in an organism together are called the genome."
Explain whether or not this statement is correct.
- 20 Explain why DNA replication must occur in a cell before mitosis.
- 21 The mass of DNA in a cell in stage 1 of mitosis is 3.0×10^{-12} g.
What is the mass of DNA in the same cell at the beginning of interphase?
Explain your answer.
- 22 Cells regulate their cell cycle very strictly. There are points in the cell cycle called checkpoints that can only be passed when everything is assessed to be normal. These checkpoints can stop working.
Suggest a consequence for the cell when these checkpoints do not work.
- 23 Name one example of a human cell that contains **no** chromosomes and explain why this is.
- 24 What are gonads? What are the male and the female gonads in humans?
- 25 What is the difference between spermatocyte I and spermatocyte II cells?
- 26 Describe three differences relating to the beginning, duration and end of gametogenesis in women and in men?
- 27 What is the second polar body?

Module 8 Heredity and Variability

Learning objectives

- Investigate the regularities of modification variability (10.2.4.1)
- Explain Mendel's two laws of genetics and how his work helps explain monohybrid and dihybrid crosses
- Explain the terms linkage and sex linkage
- Apply the cytological bases of dihybrid crosses; inheritance coupled with sex and multiple allelicism in solving problems (10.2.4.2)
- Explain irregularities in inheritance patterns as a result of cross-over (10.2.4.3)
- Compare the interaction of allelic and non-allelic genes (10.2.4.4)
- Research the theory of mutation of Hugo de Vries (10.2.4.5)
- Describe human conditions associated with chromosome number anomalies (10.2.4.6)

Gametes

All body cells except reproductive cells are called **somatic** cells. Somatic cells include cheek, liver, muscle, blood, leaf, stem and root cells.

The somatic cells in most organisms are diploid (or $2n$), which means they contain a double set of chromosomes. If these cells were to join together, the number of chromosomes in the resulting cell would be double the normal number.

In meiosis the number of chromosomes in the nucleus is halved. This means that in meiosis the diploid number is reduced to a haploid number of chromosomes. If the haploid cells formed in meiosis are capable of fusing together, they are called gametes, or sex cells.

Gametes transmit genes from one generation to another in sexual reproduction. In humans, the gametes are the sperm and egg.

The zygote normally grows (by mitosis) to form a new organism.

Gametes are haploid cells that are capable of fusion. **Fertilisation** is the union of two gametes to form a single cell called a zygote.

Genetic crosses

The mechanism of genetic crosses is best understood by the use of sample questions and their answers. Explanations for new terms are provided as they arise in each question.

Question 1

In cats, black coat (B) is dominant over white coat (b). Give the genotypes and phenotypes for the offspring of a cross involving two cats whose genotypes are (BB) and (bb).



8.1 Genetic crosses produce variations in coat colour in shorthorn cattle

Explanation of some terms

Genes are represented by letters. Usually the first letter of the dominant trait is used (e.g. B). Normally two different types of the same gene exist, i.e. a dominant version (symbolised by a capital letter, e.g. B) and a recessive version (small letter, e.g. b). These different (or alternative) versions are called alleles.

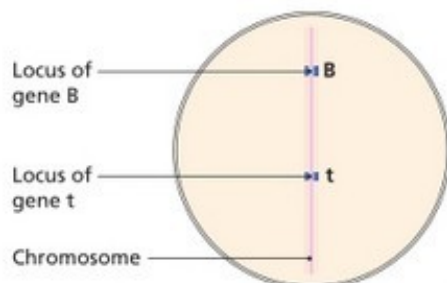
Alleles are found at the same position (or locus) on similar chromosomes.

Alleles are different (or alternative) forms of the same gene.

The locus of a gene is its position on a chromosome.

Dominant means the allele that prevents the recessive allele from being expressed.
Recessive means the allele is prevented from being expressed by a dominant allele.

When a dominant and a recessive allele occur together it is the dominant allele that works. This means that cats that are BB or Bb are both black.



8.2 The locus (plural: loci) of genes

Recessive alleles *recede* in the presence of a dominant allele. This means that cats that are Bb are black, although they contain the recessive allele (b) for white coat. For a cat to be white it has to be bb.

Normally each characteristic is controlled by a gene that has a pair of alleles. For example the gene for coat colour in the cat has two alleles: B and b. In this case the genotypes may be BB, Bb or bb.

Genotype means the genetic make-up of an organism, i.e. the genes that are present.

Phenotype means the physical make-up, or appearance, of an organism.

In Question 1 the phenotype is the coat colour of the cat (i.e. the phenotype is either black or white coat).

Genes are the instructions to the cell that help to produce the phenotype. However, genes are influenced by the environment. It is the interaction of the genes with the environment that produces the phenotype.

For instance, most people are born with the gene for the pigment melanin. However, the amount of melanin they produce will depend on their exposure to ultraviolet rays.

Genotype + environment = phenotype

Amazingly, even gender can be influenced by the environment. In some reptiles and fish the sex of the individual will vary depending on heat or light acting on the genotype. Indeed, some organisms change from male to female every second year.

The relative importance of the genotype (nature) and the environment (nurture) has been argued for generations, especially with regard to intelligence.

This **nature versus nurture** argument has not been fully settled. Present-day opinion suggests that phenotype (e.g. intelligence) is a combination of inherited genes and the upbringing of the child.

When working out genetic crosses you should realise that:

- A pair of alleles is present in the cells of an organism for each characteristic
- Only one allele for each characteristic is carried in each gamete
- As a result of gametes fusing, a pair of alleles is present in the progeny.

These rules are applied in diagram 8.3.

When writing the letters it is essential to distinguish clearly between capital and small letters.

Answer 1

B = Black coat b = White coat		
Phenotypes of parents	Black	x White
Genotypes of parents	BB	bb
Genotypes of gametes	B	b
Genotype of offspring	Bb	
Phenotype of offspring	All black	
8.3	Answer 1	

Question 2

In pea plants, green pods (G) are dominant to yellow pods (g). Show by means of diagrams the genotypes and phenotypes of the F₁ progeny that result from crossing two heterozygous plants.

Explanation of other terms

The F₁ progeny means the first generation of offspring. F₁ is short for 'first filial generation'.

Remember *homo* means 'the same as'.

Homozygous dominant = GG; homozygous recessive = gg. 'Pure breeding' is another term for homozygous.

Hetero means 'different'.

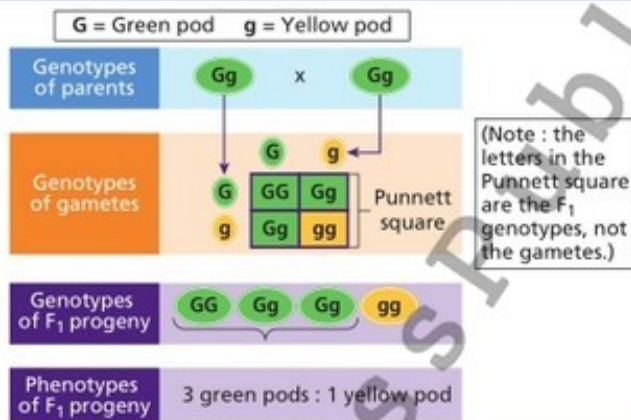
Heterozygous is also called hybrid. The genotype Gg (or gG) is heterozygous.

Progeny refers to offspring that are produced.

Homozygous means that two alleles are identical.

Heterozygous means that the alleles are different.

Answer 2



A Punnett square is a grid used to show the ratio of the genotypes of the progeny in a genetic cross.

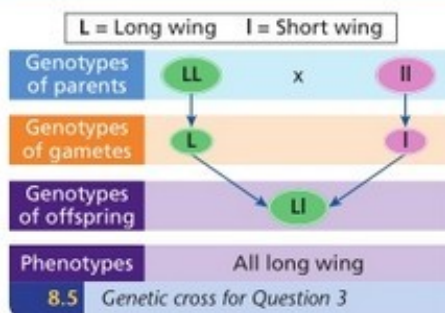
8.4 Answer 2

Question 3

In flies, long wing is dominant to short wing. A homozygous dominant fly is crossed with a homozygous recessive fly.

- What letter should represent long wing?
- Give the genotype of the homozygous dominant parent.
- State the phenotype of the homozygous dominant parent.
- Give the genotypes of all the gametes produced.
- If 100 flies are produced, how many would you expect to be:
 - Long winged
 - Homozygous dominant?


Answer 3



The cross in this case can be represented as shown above. Normally the first letter of the dominant trait is used to represent the gene.

The specific answers are:

- Long wing = L
- Genotype of homozygous dominant parent = LL
- Phenotype of homozygous dominant parent = long wings
- Gamete genotypes = L and l
- (i) Expect 100 long-winged.
(ii) Expect none of the flies to be homozygous dominant.

Question 4	Answer 4																	
<p>In the fruit fly, <i>Drosophila</i>, body colour is controlled by two alleles. The allele for grey body (G) is dominant to the allele for black body (g). If two heterozygous flies are crossed, show by diagrams that the ratio of flies with grey bodies to flies with black bodies is 3:1.</p>  <p>8.6 A grey-bodied fruit fly (<i>Drosophila</i>)</p>	<p style="text-align: center;">G = Grey body g = Black body</p> <table border="1" style="width: 100%; border-collapse: collapse;"> <tr> <td style="background-color: #003366; color: white;">Genotypes of parents</td> <td style="text-align: center;">Gg x Gg</td> </tr> <tr> <td style="background-color: #003366; color: white;">Genotypes of gametes</td> <td style="text-align: center;"> <table border="1" style="margin: auto;"> <tr> <td style="padding: 5px;">G</td> <td style="padding: 5px;">G</td> <td style="padding: 5px;">g</td> </tr> <tr> <td style="padding: 5px;">G</td> <td style="padding: 5px;">GG</td> <td style="padding: 5px;">Gg</td> </tr> <tr> <td style="padding: 5px;">g</td> <td style="padding: 5px;">Gg</td> <td style="padding: 5px;">gg</td> </tr> </table> </td> </tr> <tr> <td style="background-color: #003366; color: white;">Genotypes of progeny</td> <td style="text-align: center;">GG Gg Gg gg</td> </tr> <tr> <td style="background-color: #003366; color: white;">Phenotypes of progeny</td> <td style="text-align: center;">3 grey bodies : 1 black body</td> </tr> </table> <p>8.7 Answer 4</p>	Genotypes of parents	Gg x Gg	Genotypes of gametes	<table border="1" style="margin: auto;"> <tr> <td style="padding: 5px;">G</td> <td style="padding: 5px;">G</td> <td style="padding: 5px;">g</td> </tr> <tr> <td style="padding: 5px;">G</td> <td style="padding: 5px;">GG</td> <td style="padding: 5px;">Gg</td> </tr> <tr> <td style="padding: 5px;">g</td> <td style="padding: 5px;">Gg</td> <td style="padding: 5px;">gg</td> </tr> </table>	G	G	g	G	GG	Gg	g	Gg	gg	Genotypes of progeny	GG Gg Gg gg	Phenotypes of progeny	3 grey bodies : 1 black body
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g	Gg	gg																
Genotypes of progeny	GG Gg Gg gg																	
Phenotypes of progeny	3 grey bodies : 1 black body																	

Codominance

Normally the characteristic controlled by a dominant allele is displayed in the heterozygous genotype. The characteristic controlled by a recessive allele is normally only displayed in the homozygous recessive genotype.

Codominance is relatively rare.

Examples of codominance

- One example of codominance occurs in shorthorn cattle. In this case the genotype RR produces a red coat and the genotype rr produces a white coat. The heterozygous condition Rr gives a roan coat (patches of red and patches of white coat, as shown in diagram 8.1).
- Another example of codominance is flower colour in snapdragons. In this case RR produces red flowers, rr produces white flowers, but Rr produces pink flowers.

Codominance means that neither allele is dominant or recessive with respect to the other. Both alleles are equally expressed in the heterozygous genotype to produce an intermediate phenotype.

Question 5	Answer 5																													
<p>Flower colour in snapdragons shows codominance, i.e. the heterozygous condition (Rr) is pink. Give the phenotypes and genotypes for the progeny of the following crosses:</p> <p>(a) A white-flowered plant and a red-flowered plant</p> <p>(b) Two pink-flowered plants.</p>	<p style="text-align: center;">RR = Red flower Rr = Pink flower rr = White flower</p> <table border="1" style="width: 100%; border-collapse: collapse;"> <tr> <td></td> <td style="text-align: center;">Cross (a)</td> <td style="text-align: center;">Cross (b)</td> </tr> <tr> <td style="background-color: #003366; color: white;">Phenotypes of parents</td> <td style="text-align: center;">White flower x Red flower</td> <td style="text-align: center;">Pink flower x Pink flower</td> </tr> <tr> <td style="background-color: #003366; color: white;">Genotypes of parents</td> <td style="text-align: center;">rr x RR</td> <td style="text-align: center;">Rr x Rr</td> </tr> <tr> <td style="background-color: #003366; color: white;">Genotypes of gametes</td> <td style="text-align: center;"> <table border="1" style="margin: auto;"> <tr> <td style="padding: 5px;">r</td> <td style="padding: 5px;">R</td> </tr> </table> </td> <td style="text-align: center;"> <table border="1" style="margin: auto;"> <tr> <td style="padding: 5px;">R</td> <td style="padding: 5px;">R</td> <td style="padding: 5px;">r</td> </tr> <tr> <td style="padding: 5px;">R</td> <td style="padding: 5px;">RR</td> <td style="padding: 5px;">Rr</td> </tr> <tr> <td style="padding: 5px;">r</td> <td style="padding: 5px;">Rr</td> <td style="padding: 5px;">rr</td> </tr> </table> </td> </tr> <tr> <td style="background-color: #003366; color: white;">Genotypes of progeny</td> <td style="text-align: center;">Rr</td> <td style="text-align: center;">RR Rr Rr rr</td> </tr> <tr> <td style="background-color: #003366; color: white;">Phenotypes of progeny</td> <td style="text-align: center;">All pink flowers</td> <td style="text-align: center;">1 red flower : 2 pink flowers : 1 white flower</td> </tr> </table> <p>8.8 Answer 5</p>		Cross (a)	Cross (b)	Phenotypes of parents	White flower x Red flower	Pink flower x Pink flower	Genotypes of parents	rr x RR	Rr x Rr	Genotypes of gametes	<table border="1" style="margin: auto;"> <tr> <td style="padding: 5px;">r</td> <td style="padding: 5px;">R</td> </tr> </table>	r	R	<table border="1" style="margin: auto;"> <tr> <td style="padding: 5px;">R</td> <td style="padding: 5px;">R</td> <td style="padding: 5px;">r</td> </tr> <tr> <td style="padding: 5px;">R</td> <td style="padding: 5px;">RR</td> <td style="padding: 5px;">Rr</td> </tr> <tr> <td style="padding: 5px;">r</td> <td style="padding: 5px;">Rr</td> <td style="padding: 5px;">rr</td> </tr> </table>	R	R	r	R	RR	Rr	r	Rr	rr	Genotypes of progeny	Rr	RR Rr Rr rr	Phenotypes of progeny	All pink flowers	1 red flower : 2 pink flowers : 1 white flower
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Genotypes of progeny	Rr	RR Rr Rr rr																												
Phenotypes of progeny	All pink flowers	1 red flower : 2 pink flowers : 1 white flower																												

Pedigree studies

Question 6	Answer 6
<p>In humans, the ability to produce the skin pigment melanin is controlled by a dominant allele (N). Lack of pigment (albinism) is controlled by the recessive allele (n). The pedigree for a family is represented below.</p> <div style="border: 1px solid black; padding: 5px; margin: 10px 0;"> = Male (not albino) = Male albino = Female (not albino) = Female albino </div> <p>8.9 Family pedigree for skin colour</p> <p>(a) Give the genotypes of persons 1, 2 and 5. (b) Could person 6 be homozygous dominant? Give a reason for your answer. (c) How many children had the parents 1 and 2? (d) Give all the possible genotypes for person 4.</p>	<p>N = skin with pigment, n = Albino</p> <p>(a) Persons 1 and 2 have skin with pigment but their child (person 5) is an albino, nn. This means that persons 1 and 2 must both be Nn. Person 5 must be nn, i.e. albino.</p> <p>(b) Person 6 cannot be homozygous dominant. If person 6 was NN then all his children would have skin with pigment. However, one of his children (person 7) is albino, nn. Thus, person 6 must be Nn.</p> <p>(c) Parents 1 and 2 had three children (i.e. persons 3, 4 and 5).</p> <p>(d) Person 4 could be NN or Nn.</p> <div style="background-color: #fff9c4; padding: 10px; margin-top: 10px; border: 1px solid #ccc;"> <p>A pedigree is a diagram showing the genetic history of a group of related individuals.</p> </div>

Summary of the ratios of genetic crosses

Note: In the first three crosses (or rows) below it is assumed that B (black coat) is dominant over b (white coat). In the final cross the assumption is that RR = red coat, Rr = roan coat and rr = white coat.

Ratio	Example	Explanation
1 : 0 or 100%	All offspring are black or 84 out of 84 of the offspring are black	BB × BB BB × Bb BB × bb
1 : 1 or 50% : 50%	Equal numbers of black and white offspring were produced or 164 black and 159 white	Bb × bb
3 : 1 or 75% : 25%	35 black and 11 white or 124 black and 41 white	Bb × Bb
1 : 2 : 1 or 25% : 50% : 25%	19 red, 40 roan and 22 white offspring	Rr × Rr (where alleles show incomplete dominance)

Sex determination

The nucleus of each human somatic (or non-gamete-forming) cell has 46 chromosomes (i.e. $2n = 46$). These 46 chromosomes consist of 44 non-sex chromosomes called **autosomes** and two sex chromosomes.

The autosomes control features that are independent of whether a person is male or female. Examples of such gender-neutral features are skin colour, number of arms, production of saliva and digestive enzymes.

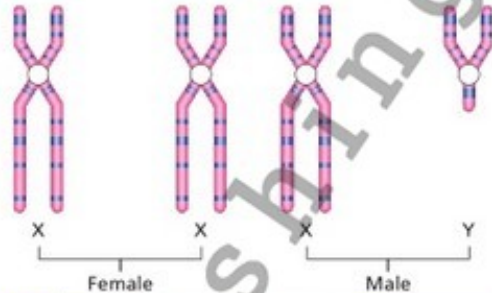


8.10 Human sex chromosomes: note that the X chromosome (left) is much larger than the Y chromosome

The two sex chromosomes are called the X and Y chromosomes. They contain genes that control gender in most species. The X chromosome is longer than the Y chromosome.

Humans

In humans every individual somatic cell nucleus should have two sex chromosomes. If these are XX the individual is female; if they are XY the individual is male.



8.11 The sex chromosomes in humans

The arrangement of XX for females and XY for males has the following two consequences:

- It is the male who determines the sex of the child
- The ratio of male to female births should be equal (1:1).

Question 7	Answer 7				
<p>Show using diagrams why the father determines the sex of a child in humans.</p>	<div style="display: flex; justify-content: space-around;"> <div style="text-align: center;"> <p>Female</p> <p>XX</p> <p>↓</p> <p>Egg X</p> </div> <div style="text-align: center;"> <p>x</p> </div> <div style="text-align: center;"> <p>Male</p> <p>XY</p> <p>↓</p> <p>Sperm X Sperm Y</p> </div> </div> <div style="text-align: center; margin-top: 10px;"> <table border="1" style="margin-left: auto; margin-right: auto;"> <tr> <td style="background-color: #90EE90;">XX</td> <td style="background-color: #FFD700;">XY</td> </tr> <tr> <td style="text-align: center;">Female child</td> <td style="text-align: center;">Male child</td> </tr> </table> </div> <p>8.12 Answer 7</p> <p>The female always donates an X chromosome (in the egg) to her children. The chance of a boy or a girl being formed depends on the sex chromosome in the sperm.</p> <ul style="list-style-type: none"> • If a sperm containing an X chromosome fertilises the egg, a girl is produced. • If the fertilising sperm carries a Y chromosome, a boy is formed. <p>This means that the sex of a child is determined by the genotype of the sperm that fertilises the egg, i.e. it is the father's sperm that determines the sex of the child.</p>	XX	XY	Female child	Male child
XX	XY				
Female child	Male child				

Other species

The pattern of sex determination in some species is the reverse of that in humans. For example, in birds, butterflies and moths males are XX and females are XY.

Ratio of male to female births in humans

Diagram 8.12 shows that the chance of an XX (girl) or XY (boy) offspring being formed is 50% (i.e. 1:1). This is because equal numbers of sperm containing an X chromosome and sperm containing a Y chromosome are produced. Therefore, if a woman is pregnant it is equally likely that her child will be a boy or a girl.

The work of Gregor Mendel

Gregor Mendel is known as the 'father of genetics'. He was born in Austria in 1822 and became an Augustinian monk at the age of 21. Having twice failed his teacher's qualifying examination, he turned to the study of the edible or garden pea plant.

Mendel carried out numerous experiments on garden pea plants. He investigated the inheritance of seven characteristics of these plants, such as stem height, flower colour and seed shape.

These experiments involved removing the pollen-producing structures (called anthers) from some flowers and transferring pollen from other flowers to the treated flowers by hand. The treated flowers were then covered with bags to prevent any more pollen from reaching them. The seeds that formed were collected and grown in carefully labelled containers. The appearance (phenotype) of the resulting plants was studied and recorded.

The success of Mendel's work was largely due to two main features.

- He only studied features (or characteristics) that displayed two forms (or traits). For instance, the plants were either tall or small and the pods were either green or yellow.
- He counted the number of plants with each type of trait. He was able to detect mathematical ratios such as 1:1 or 3:1 from the numerical data he obtained.

Mendel's research was carried out around 1860 and resulted in two basic laws of inheritance (called Mendel's first and second laws). His results were ignored until 1900, when a number of researchers discovered the significance of his studies.



8.14 Mendel studied the production of pea characteristics, such as round or wrinkled peas



Despite the ratio being equal in theory, the numbers of male and female births is not equal. Large-scale worldwide studies suggest that boys are more likely to be born than girls (the figures are close to 106:100). This imbalance may occur to compensate for the fact that more males die early in life than females. The exact reason for the imbalance is not known.



8.13 Gregor Mendel (1822–1884)

The law of segregation (Mendel's first law) states that:

- Inherited characteristics are controlled by pairs of alleles.
- These alleles segregate (or separate) from each other at gamete formation, with only one member of the pair being found in each gamete.

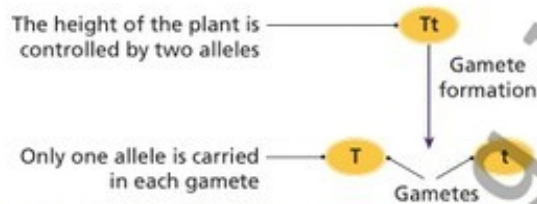
Mendel's first law

Mendel's first law is called **the law of segregation**.

Example of the law of segregation

If, for a species of plant, T = tall and t = small, the height of the plant is controlled by a pair of alleles, e.g. TT, Tt or tt.

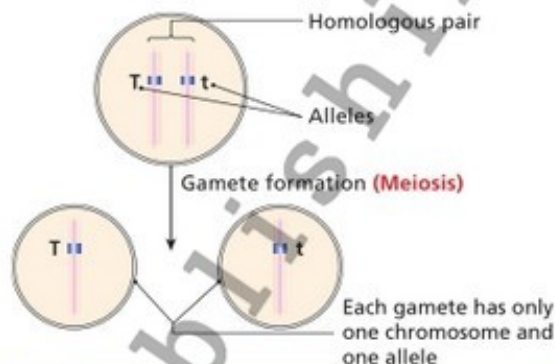
When gametes are formed, only one allele can enter each gamete (the alleles segregate or separate). This is shown in diagram 8.15.



8.15 Example of Mendel's first law of segregation

Chromosomal basis of Mendel's first law

- 1 In diploid organisms, chromosomes occur in matching pairs (called homologous pairs).
- 2 Pairs of alleles occupy the same position (locus) on a homologous pair.
- 3 During meiosis, homologous chromosomes separate and go into different cells.
- 4 As a result, pairs of alleles also separate (see diagram 8.16).



8.16 The behaviour of chromosomes due to Mendel's first law

Monohybrid and Dihybrid crosses

A monohybrid cross involves the study of a single characteristic.

Examples of monohybrid crosses involve features such as eye colour, seed shape or coat colour. Each characteristic can display two variations or phenotypes. This means that a characteristic such as eye colour can display two phenotypes, such as brown eyes or blue eyes.

All the examples of genetic crosses given in this module so far are monohybrid crosses.

A dihybrid cross involves the study of two characteristics.

For example, Question 8 is a dihybrid cross because it involves two characteristics: plant size (tall or small) and pod colour (green or yellow).

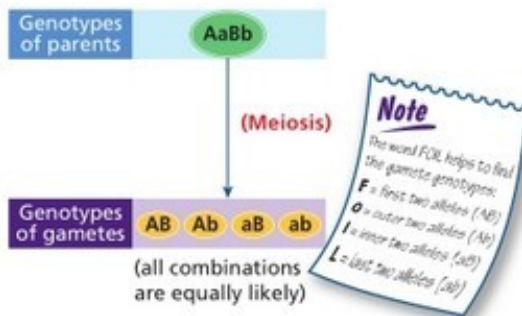
The law of independent assortment states that:

- When gametes are formed ...
- either of a pair of alleles ...
- is equally likely ...
- to combine with either of another pair of alleles.

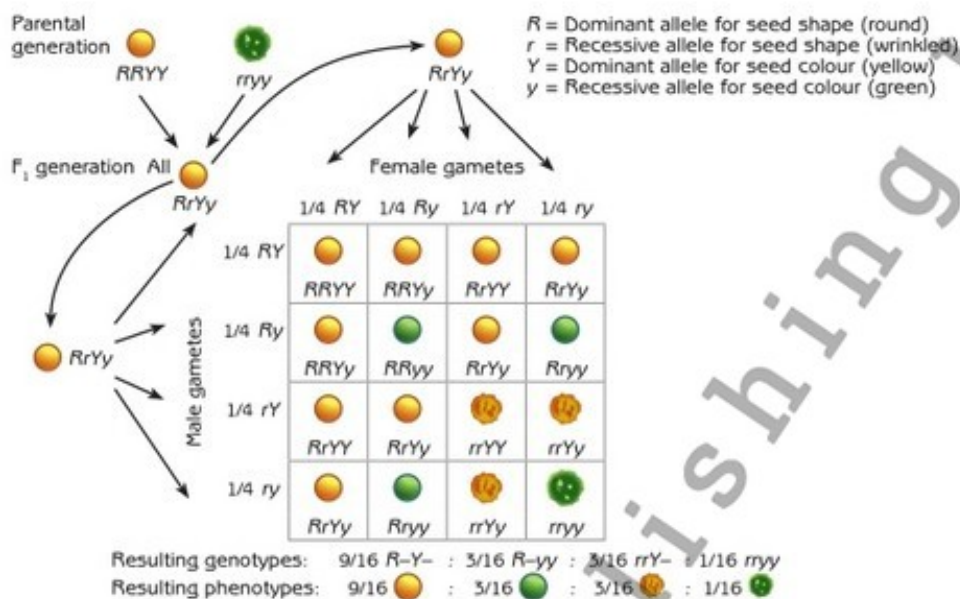
Mendel's second law

Mendel carried out a range of dihybrid crosses. Having analysed his results, he formulated his second law, the law of independent assortment.

The second law means that in an organism with the genotype AaBb either of the As can combine with either of the Bs to form gametes. As a result, the four gamete types shown in diagram 8.17 are equally likely to be formed.



8.17 Example of Mendel's second law of independent assortment

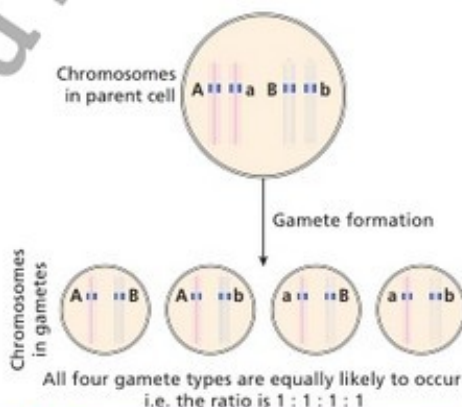


8.18 Example of a dihybrid cross

Chromosomal basis of Mendel's second law

As alleles are located on homologous chromosomes, Mendel's second law can be restated as:

- At gamete formation ...
- either of a pair of homologous chromosomes ...
- is equally likely ...
- to combine with either chromosome of a second homologous pair.



Examples of dihybrid crosses

8.19 The behaviour of chromosomes due to Mendel's second law

Question 8

In pea plants, tall plant (T) is dominant over small plant (t). In addition, green pod (G) is dominant over yellow pod (g).

A tall plant with green pods (homozygous for both traits) is crossed with a small plant with yellow pods.

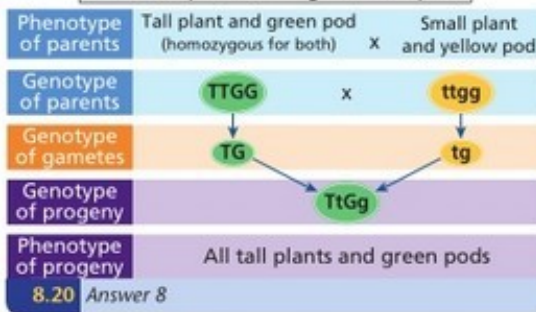
- (a) Why is this a dihybrid cross?
- (b) Show using diagrams the genotypes and phenotypes of the progeny of this cross.

Answer 8

(a) This is a dihybrid cross because two characteristics are studied, i.e. plant height and pod colour.

(b)

T = Tall plant G = Green pod
t = Small plant g = Yellow pod



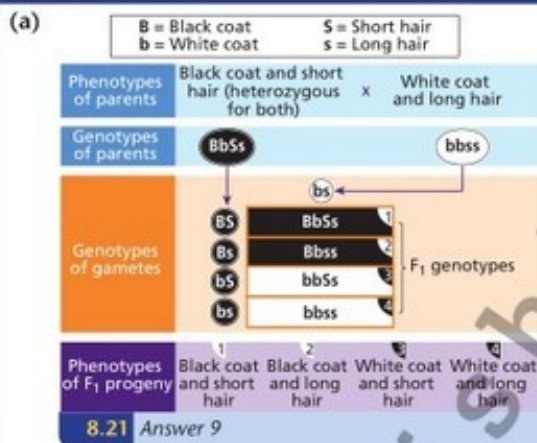
8.20 Answer 8

Progeny genotypes: TtGg. Progeny phenotypes: all tall plants and green pods.

Question 9

In guinea pigs, black coat (B) is dominant to white coat (b). Also short hair (S) is dominant to long hair (s).

- (a) Show the genotypes and phenotypes of the F₁ progeny for a cross involving a black-coated, short-haired guinea pig (heterozygous for both traits) and a white-coated, long-haired guinea pig.
- (b) State the expected ratio of the offspring.

Answer 9


- (b) The offspring are expected to occur in equal numbers (i.e. the ratio is 1:1:1:1).

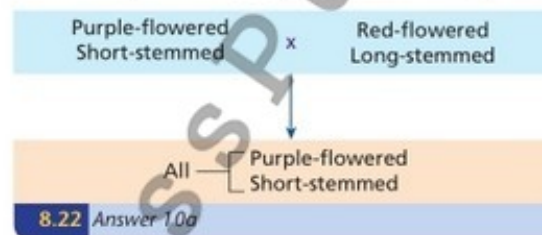
Question 10

A homozygous purple-flowered, short-stemmed plant was crossed with a red-flowered, long-stemmed plant. All the F₁ offspring were purple-flowered with short stems.

- (a) State the dominant and recessive traits.
- (b) Explain, using diagrams, why the F₁ plants all had the same phenotypes.
- (c) Give the expected phenotype ratios if an F₁ plant is selfed.

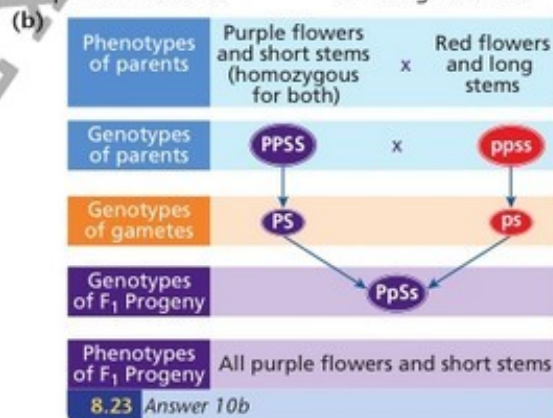
Answer 10

- (a) This cross may be summarised as shown in diagram 8.22:



The dominant traits appear in the F₁ generation. This means the dominant traits are purple-flowered and short-stemmed. (Red-flowered and long-stemmed are the recessive traits.) Normally the first letter of the dominant trait is used to represent the gene. The alleles in this cross should be represented as:

P = Purple-flowered S = Short-stemmed
p = Red-flowered s = Long-stemmed



- (c) Selfing means you cross a genotype with the same genotype. In this case the F_1 genotype is $PpSs$ and this is crossed with another plant with the genotype $PpSs$. If two F_1 offspring are crossed the organisms produced are called the F_2 progeny (i.e. the second filial generation).

Phenotypes of parents	Purple flower short stem	x	Purple flower short stem		
Genotypes of parents	$PpSs$	x	$PpSs$		
Genotypes of gametes	PS	Ps	pS		
	PS	PPSS	PPSs	PpSS	PpSs
	Ps	PPSs	PPss	PpSs	Ppss
	pS	PpSS	PpSs	ppSS	ppSs
ps	PpSs	Ppss	ppSs	ppss	
Phenotypes of F_2 progeny	9 Purple flowers and short stems 3 Purple flowers and long stems 3 Red flowers and short stems 1 Red flowers and long stems				
8.24 Answer 10c					

Expected ratios arising from Mendel's second law

In a dihybrid cross where one parent is heterozygous for both characteristics and the second parent is homozygous recessive (e.g. $BbSs \times bbss$), if the offspring are produced in the ratio 1:1:1:1 then independent assortment has occurred.

Also if both parents are heterozygous (e.g. $PpSs \times PpSs$) and the offspring are in the ratio 9:3:3:1 then, again, independent assortment has occurred.

Chromosome diagrams

The **locus** of a gene is its position on a chromosome. The locus (plural: loci) of a gene is shown in diagram 8.25. Alleles occupy similar loci on homologous chromosomes, as shown in diagram 8.26.

Linkage

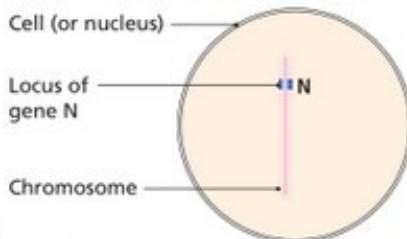
Linked genes tend to be passed on together (forming a linkage group) to the next generation, i.e. they do **not** show independent assortment.

Linkage means that genes are located on the same chromosome.

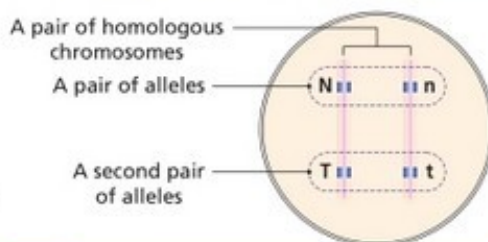
Example of linked genes

In diagram 8.27:

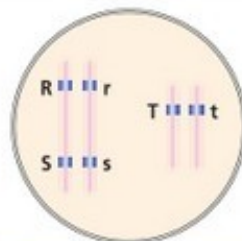
- There are four chromosomes
- The alleles R and S are linked, as are the alleles r and s
- Nothing is linked to the alleles T and t
- R and r (along with S and s as well as T and t) are alleles
- R and S are *not* alleles, nor are R and s nor R and T.



8.25 The locus of a gene



8.26 Alleles on chromosomes



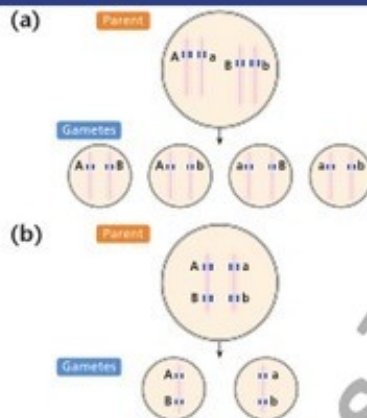
8.27 A simple chromosome diagram showing linkage

Question 11

Draw simple chromosome diagrams to illustrate the following cells. In each case show the gametes that might be produced.

- (a) The genes are not linked and the genotype is AaBb.
 (b) The genes are linked (A to B and a to b) and the genotype is AaBb.

Answer 11



8.28 Answer 11

Note that in Answer 11:

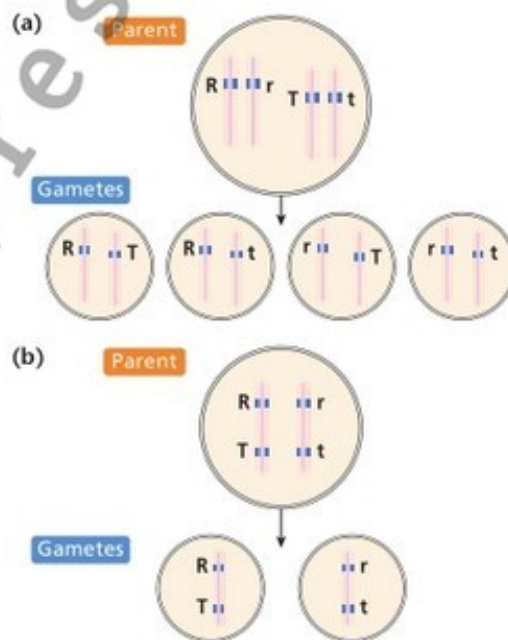
- (a) The four gamete types are equally likely, i.e. the ratio is 1:1:1:1.
 (b) The genes are linked. In this case only two types of gametes are formed, i.e. the only gametes are (AB) and (ab). This is a contradiction of the law of independent assortment. For this reason we can say that linkage **contradicts** Mendel's second law of independent assortment.

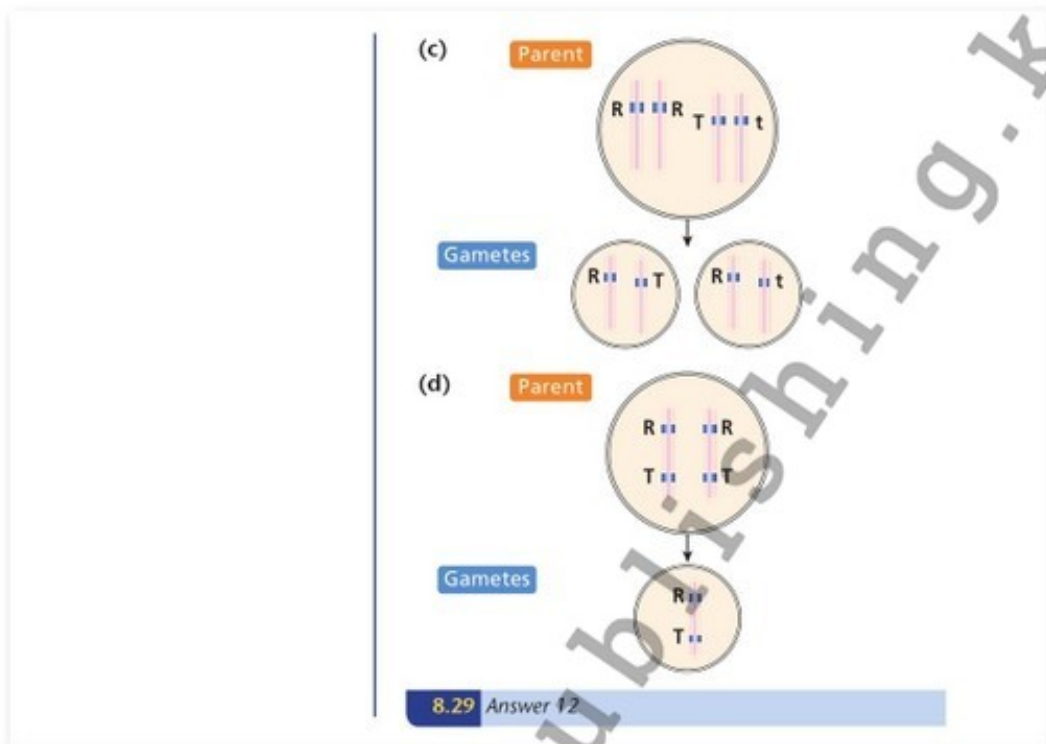
Question 12

Draw simple chromosome diagrams to show each of the following cells. In each case indicate the gametes that each cell might produce.

- (a) The genotype is RrTt, the genes are not linked.
 (b) The genotype is RrTt, the genes are linked (R to T and r to t).
 (c) The genes are not linked; the cell is homozygous for R and heterozygous for T.
 (d) The genes are linked and the cell is homozygous dominant for both genes.

Answer 12





The ratio of offspring in linked crosses

In parts (a) and (b) of Question 12 the parents have the same genotypes (i.e. both are RrTt). However, different types of gametes are formed in each case.

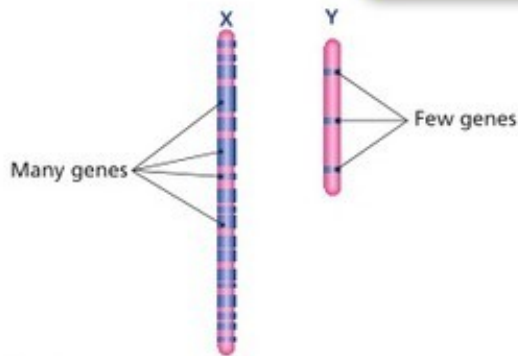
The ratios of the **genotypes of the gametes** produced by linked crosses are different from those produced in non-linked crosses. This results in linked crosses producing different ratios of **offspring** than might otherwise be expected.

Sex linkage

The sex chromosomes in humans are the X and Y chromosomes. The X chromosome carries a large number of genes. The Y chromosome is much shorter than the X and carries very few genes.

Sex linkage means that a characteristic is controlled by a gene on a sex (or X) chromosome.

? The main gene isolated on the Y chromosome is the SRY gene. This stands for the Sex-determining Region of Y. It, and a small number of other genes on the Y chromosome, control the development of the testes. These genes are thought to control maleness.



8.30 The sex chromosomes

Sex-linked characteristics are also said to be X-linked. Examples of sex-linked characteristics are:

- Colour blindness
- Haemophilia (inability to clot blood)
- Duchenne muscular dystrophy (where the muscles waste away, resulting in early death)
- Eye colour in *Drosophila* (the fruit fly, often used in genetics experiments).

All these characteristics are controlled by genes, or alleles, located on the X chromosome. In males there is no corresponding allele on the Y chromosome.

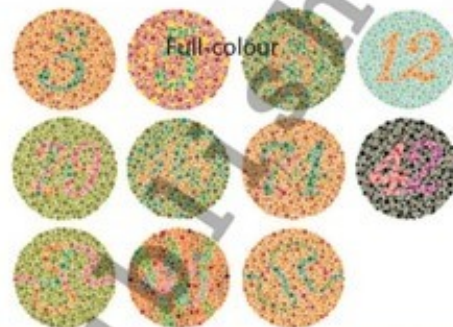
In sex-linked characteristics, the recessive phenotype is more likely to occur in males, i.e. males suffer more often from sex-linked characteristics (as shown in the following examples).

Examples of sex-linked characteristics

Colour blindness

Normal individuals can detect three colours of light (red, green and blue). The allele for full-colour vision (C) is dominant. Colour blindness (c) usually means an inability to distinguish red from green.

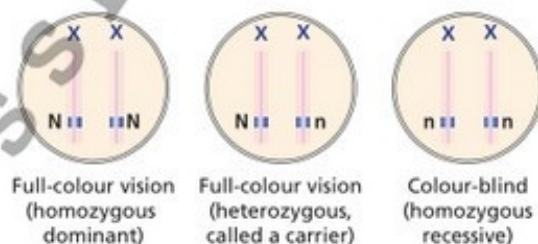
The gene for colour vision is located on an X chromosome.



8.31 Test for colour blindness: a person with full-colour vision can distinguish the number or pattern in each disc

Females

Females can have three distinct genotypes with respect to colour vision. These are shown in diagram 8.32. Note that these genotypes can be represented as shown in the diagram or as $XNXN$ or $XXNn$ or $XnXn$. For a female to be colour-blind, she needs **two** copies of the recessive allele (n). As recessive alleles are usually relatively scarce, it is rare to have two recessives.



8.32 The alleles for colour vision in females

Males

Males have only one allele for colour vision. This is on the X chromosome. The Y chromosome has no allele for colour vision.

This means there are only two genotypes for males, as shown in diagram 8.33.

These genotypes can also be given as XNY or XYn .

Males only need **one** recessive allele in order to be colour-blind. This means that males are more likely to be colour-blind than females. Most of these males have some degree of colour vision. Complete colour blindness is very rare.



8.33 The alleles for colour vision in males

Haemophilia

Haemophilia is a bleeding disorder caused by the lack of a particular blood protein. Haemophiliacs suffer from frequent bleeding, often into the joints. Without treatment, some haemophiliacs may bleed to death after a small cut.

Haemophilia is caused by a gene located on the X chromosome. The allele (N) for the production of the clotting protein is dominant. The recessive allele (n) does not carry the correct genetic code for the production of the protein.

As with all sex-linked traits, haemophilia is more common in males (0.01%) than in females, where it is extremely rare. Males only need a single copy of the recessive allele (n) to be haemophiliac, but females need two copies of the recessive allele.



8.34 A baby with haemophilia: notice the bruising caused by this bleeding disorder

Summary of the ratios of (dihybrid) genetic crosses

Note: In the crosses below it is assumed that B (black coat) is dominant over b (white coat) and L (long tail) is dominant over l (short tail).

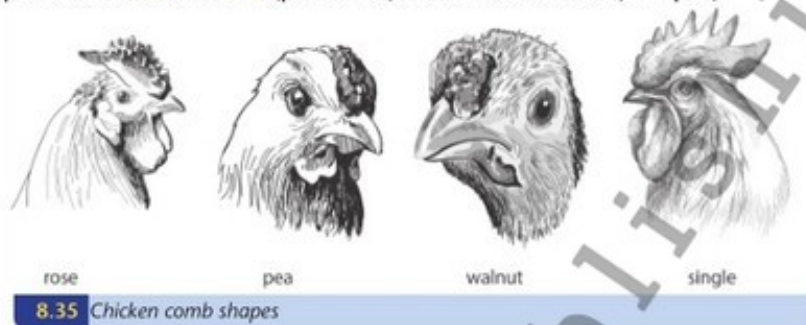
Ratio	Example	Explanation
1 : 0 or 100%	All offspring were black-coated with long tails	BBLL × bbll or BBll × bbLL (other explanations exist, but the two shown are the most commonly asked)
1 : 1 : 1 : 1 or 25% : 25% : 25% : 25%	All four offspring genotypes were formed in equal numbers or Black coat, long tail = 18 Black coat, short tail = 17 White coat, long tail = 19 White coat, short tail = 17	BbLl × bbll (where the genes are not linked)
1 : 1	Equal numbers of black-coated, long-tailed and white-coated, short-tailed or Black coat, long tail = 76 White coat, short tail = 75	BbLl × bbll (where the genes are linked)
9 : 3 : 3 : 1	Black coat, long tail = 176 Black coat, short tail = 62 White coat, long tail = 59 White coat, short tail = 20	BbLl × BbLl

Epistasis

Epistasis is where one gene affects the expression of another, different, gene. There are many ways that this can happen, and epistasis can be dominant or recessive.

One example of this is the shape of the comb in some birds. The comb is a crest on top of the head of birds like turkeys and domestic chickens. In domestic chickens, there are four comb shapes shown in diagram 8.35.

One gene has a pair of alleles **A** and **a**. **A** (rose comb) is dominant over **a** (non-rose). The other gene has a pair of alleles **B** and **b**. **B** (pea comb) is dominant over **b** (non-pea).



If rose and pea are present together (alleles **A** and **B**), the phenotype is walnut. If neither pea nor rose are present (**aabb** only), then the phenotype is single.

For example, genotype **AabB** would be walnut, **Aabb** would be rose, **aaBB** would be pea, etc.

Other factors influencing variation

Not all characteristics are controlled by genes. Most characteristics of organisms are determined by a combination of genetic inheritance and environmental influence. For example, the height of a person will be determined by genes that are inherited from parents, but also factors such as nutrition during childhood, or even certain diseases. It is possible, for example, that identical twins may not look exactly identical due to these and other factors. In fact, most human characteristics are influenced by the environment. Other examples include the colour of flowers and the number of leaves on a plant. The basic flower colour will be determined by genetics but can be influenced by the mineral content of the soil. The number of leaves may be influenced by water availability or amount of sunlight.

Non-nuclear inheritance

Most of the DNA (and genes) in a cell is located in the nucleus. However, non-nuclear or extra-nuclear genes are present as small circles of DNA in mitochondria and chloroplasts. Both of these organelles reproduce by themselves and pass on their genes to the resulting organelles.



In plants, mitochondria and chloroplasts are normally passed on to the next generation in the cytoplasm of the egg. Pollen does not contain these organelles.

In animals, mitochondria are in the tail of the sperm. Only the head of the sperm joins with the egg (the tail stays outside the egg). This means that mitochondria from the sperm are not passed on to the zygote.

Mitochondria and chloroplasts are said to follow a maternal line of inheritance, i.e. they are inherited from the female in the cytoplasm of the egg.

For example, mitochondrial DNA (mtDNA) in humans is inherited only from the mother. A number of rare human disorders are inherited only from the mother because they are controlled by non-nuclear genes located on mtDNA. These disorders normally involve a lack of energy (ATP) and affect systems with high energy demands, such as the muscular and nervous systems.



In groups, research Hugo de Vries theory of mutation and prepare a short PowerPoint presentation.

Karyotyping

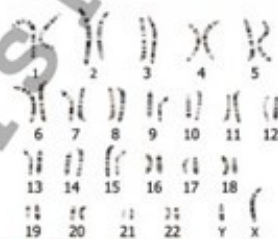
A karyotype is a full set of chromosomes of an individual which are arranged in sequence. It is a procedure that evaluates the number and structure of a person's chromosomes in order to detect abnormalities. A karyotype may be used to diagnose genetic diseases and some birth defects, such as Down syndrome, or leukaemia and lymphoma.

Human Karyotyping Activity.

Your teacher will provide you with cut-out images of a complete set of chromosomes in a somatic cell. Pair the sets of cells using:

- size
- placement of the centromere
- banding pattern

Place pairs in order of size [and number the pairs] with the exception of the sex chromosomes which should be placed last.



8.36 Human karyotype

Discuss these questions with your partner.

- How many individual chromosomes are present in your karyotype?
- How many chromosomes would be present in each body (somatic) cell?
- How many chromosomes would be present in each sex (gamete) cell?
- Does your karyotype represent a female or a male?

Questions on Module 8

- 1 Explain what is meant by:
 - (a) Somatic cells
 - (b) Gametes
 - (c) Diploid
 - (d) Haploid
 - (e) Fertilisation.
- 2 Name the process in each case responsible for converting: (a) Diploid cells into haploid cells (b) Haploid cells into diploid cells.
- 3 In corn plants, yellow seed (Y) is dominant to green seed (y). A pure breeding (homozygous) yellow-seeded corn plant is crossed with a green-seeded corn plant.
 - (a) Using diagrams show the F_1 genotype and phenotype.
 - (b) From your diagrams state:
 - (i) The genotypes of the parents
 - (ii) The genotypes of the gametes
 - (iii) The genotype and phenotype of the F_1 generation.
 - (c) State the genotypes of the gametes that could be produced by the F_1 progeny.
- 4 In humans, the gene for brown eyes (B) is dominant to that for blue eyes (b). If both parents are heterozygous for the trait:
 - (a) Give the genotypes of the parents.
 - (b) Give the genotypes of the gametes produced.
 - (c) Give the possible genotypes and phenotypes of the children.
 - (d) What percentage of the children would you expect to be:
 - (i) Homozygous dominant
 - (ii) Homozygous recessive
 - (iii) Heterozygous
 - (iv) Homozygous?
- 5 In a flower, red petal is dominant to white petal. A plant, homozygous for the dominant allele, is crossed with a plant with white petals.
 - (a) Suggest suitable symbols for the two alleles.
 - (b) Give the genotypes of each parent.
 - (c) Give the genotypes of the gametes produced by each parent.
 - (d) Give the genotype of the F_1 progeny.

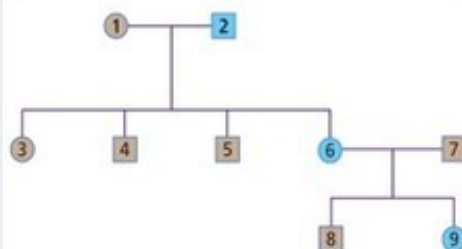
- 6 In peas, green seed (G) is dominant to yellow seed (g). A pea plant, pure-breeding for green seed, is crossed with a yellow-seeded plant. Find the genotypes and phenotypes produced in this cross.
- 7 In cats, black coat (B) is dominant to white coat (b). The genotype of the male cat is heterozygous. The female cat has the phenotype white coat. Half of the progeny of these two cats are homozygous recessive.
- (a) Explain the meaning of each of the six underlined terms.
- (b) Show this cross diagrammatically.
- (c) Use each of the underlined terms at least once to label your diagrams.
- (d) What is the phenotype of the kittens that are not homozygous recessive?
- 8 In humans, normal skin colour (N) is dominant to albinism (n).
- (a) State the phenotypes of individuals who are:
- Homozygous dominant
 - Homozygous recessive
 - Heterozygous.
- (b) Give the genotypes of the three individuals at (a) above.
- (c) Show by diagrams how two parents with skin with pigment can have an albino child.
- (d) An albino man has a daughter with skin with pigment. The daughter marries another albino man. Show by diagrams the percentage chance of their child having skin with pigment.
- 9 In a species of organism the allele D is dominant over the allele d. Diagram 8.37 shows the genotypes of two members, A and B, of the species of organism.



8.37 Different genotypes

- (a) Do these two organisms have the same phenotype? Explain your answer.

- (b) Give the possible genotypes of the gametes produced by each organism.
- (c) If A was crossed with B, could any of the offspring have the genotype dd? Explain your answer.
- 10 In cucumber plants, the character non-bitter fruit (n) is recessive to bitter fruit (N). If two heterozygous plants are crossed, show using diagrams that the ratio of bitter to non-bitter fruit is 3:1.
- 11 In shorthorn cattle, coat colour shows codominance; the heterozygous condition is roan. Show the genotypes and phenotypes of the progeny for each of the following crosses:
- A red male and a white female
 - Two roan parents.
- 12 Coat colour in collies shows codominance. The gene for black coat (B) shows equal dominance with the gene for white coat (b). The heterozygous condition is called mixed coat.
- (a) State the genotypes of a:
- Black collie
 - White collie.
- (b) State the genotypes and phenotypes of the F_1 offspring that would result from crossing a black collie and a white collie.
- (c) State the genotypes and phenotypes of the progeny of a cross between two mixed-coated collies.
- 13 Blue eyes are recessive to brown eyes. Diagram 8.38 shows the pedigree for eye colour in a family. Answer the questions that follow, using B to represent the dominant allele.



8.38 Pedigree diagram

- (a) What is the relationship between persons:
 (i) 3 and 5
 (ii) 3 and 7
 (iii) 1 and 8?
- (b) What are the genotypes of persons 1, 6 and 8?
- (c) If person 8 married a brown-eyed female, could they have any blue-eyed children? Explain your answer by means of diagrams.
- 14 Name the sex chromosomes in human:
 (a) Males (b) Females (c) Eggs
 (d) Sperm.
- 15 'A mother's chromosomes cannot determine the gender of her child.' Is this statement valid? Support your answer by including a diagrammatic cross.
- 16 Explain, giving an example in each case, what is meant by:
 (a) Allele
 (b) Genotype
 (c) Phenotype
 (d) Homozygous
 (e) Heterozygous
 (f) Dominant
 (g) Recessive
 (h) Codominance
 (i) Autosomes.
- 17 Some of the results of Mendel's crosses carried out on pea plants are given below.

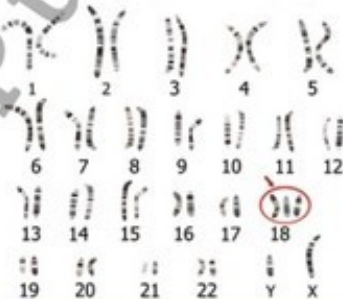
Traits	Original cross	F ₁ progeny
Seed form	Round × wrinkled	Round
Seed colour	Yellow × green	Yellow
Flower position	Axial × terminal	Axial
Pod form	Inflated × constricted	Inflated

- (a) Use suitable letters to show the genotypes of each of the following:
 (i) A green-seeded plant
 (ii) Homozygous dominant for seed form
 (iii) Heterozygous for flower position
 (iv) Homozygous recessive for pod form.
- (b) Give the phenotypes for the plants at (ii) and (iv) above.
- (c) Give the genotypes of the gametes that could result from (ii) and (iv) above.

- (d) Two plants, each with inflated pods, were crossed: 75% of the progeny had inflated pods and 25% had constricted pods. What does this indicate about the genotypes of the parent plants?
- (e) Show the results you would expect for the F₂ generation for the original cross involving seed form.
- 18 (a) State Mendel's first law of segregation.
 (b) Show how this law applies to a cell with the genotype Tt.
- 19 State Mendel's second law of independent assortment.
- 20 In snapdragons, flower colour can be red (RR), white (rr) or, in the heterozygous condition, pink. Also, tall (T) is dominant over dwarf (t).
 A dwarf, red-flowered snapdragon plant was crossed with a homozygous tall, white-flowered snapdragon plant. State:
 (a) The genotypes of the parents
 (b) The genotypes of the gametes
 (c) The possible genotypes and phenotypes of the offspring.
- 21 In shorthorn cattle, the colours red and white show incomplete dominance. In addition, the polled condition (without horns) is dominant over the horned condition. Show the results of a cross between a horned roan male and a polled white female. (**Note:** this question involves two crosses).
- 22 Draw simple chromosome diagrams to illustrate each of the following, given that the allele A is dominant over a and that the allele B is dominant over b.
 (a) The genes for A and B are not linked and the organism is heterozygous for both genes.
 (b) The genes are linked, A to B and a to b, and the organism is heterozygous for both genes.
 (c) The genes are not linked and the organism is heterozygous for A and homozygous for B.
- 23 Draw a large diagram of a cell nucleus with two pairs of chromosomes, each pair of chromosomes to be visibly distinguishable from the other pair. Indicate on the chromosomes the alleles A/a and R/r so that the nucleus is heterozygous for both genes and the genes are not linked.

- 24 An organism of genotype SsTt was crossed with one of genotype sstt. Show the progeny genotypes that could be produced if:
- The genes were linked
 - The genes were not linked. In each case, indicate the expected ratio of each genotype.
- 25
- What is meant by sex linkage?
 - Give two examples of sex-linked traits in humans.
- 26 Colour blindness is caused by a gene located on an X chromosome. Full-colour vision (C) is dominant to colour blindness (c). Two parents with full-colour vision have a colour-blind son.
- Give the genotypes of both parents and their son.
 - Did the son inherit the recessive allele from the mother or the father?
 - What is the chance of this couple having a colour-blind daughter? Explain your answer.
- 27 In humans, red-green colour blindness is a sex-linked trait. A colour-blind man and his wife have two sons. One of the sons is colour-blind and the other is not.
- What is the genotype of the mother? Outline your reasoning.
 - If a future child of this couple is male, what is the chance that he will be colour-blind? Outline your reasoning.
- 28 Explain the meaning of the following terms:
- Linked genes
 - Locus.
- 29
- Distinguish between nuclear and non-nuclear DNA.
 - State two places where non-nuclear genes may be found.
 - 'Non-nuclear DNA is only inherited from our mothers.' Explain why this statement is true for humans.

- 30 Read the information on epistasis on page 112 and use this to answer these questions.
- What is the phenotype of comb in a bird with genotype:
 - AABb
 - aAbb
 - Give the genotype of a bird that has a single comb.
 - A bird with a rose comb is crossed with a bird with a pea comb. Is it possible to get a bird with a single comb in the offspring? Explain your answer.
- 31 The height of an adult human is controlled by genetics but influenced by the environment. Give two other human characteristics that are influenced by the environment and describe what environmental factor affects them.
- 32 The drawing shows the chromosomes from a human cell.



8.39 Human karyotype

State and explain whether this cell is normal and whether it comes from a male or female.

Module 9 Evolution and Variation

Learning objectives

- Describe various chromosomal disorders in humans (10.2.4.6)
- Explain the interaction between heredity, variability and evolution (10.2.6.1)
- Analyse factors influencing the evolution process (10.2.6.2)
- Analyse different forms evidence of evolution (10.2.6.3)
- Describe the structure and stages of formation of life on Earth (10.1.1.1)
- Compile and interpret phylogenetic maps (10.1.1.2)
- Compare the principles of different forms of phylogenetic maps [cladograms and phylogenetic trees] (10.1.1.3)
- Identify ways of speciation (10.2.6.4)
- Classify the mechanisms of speciation (10.2.6.5)
- Investigate ways to improve crop plants and animal breeds through selective breeding and cross-breeding (10.2.5.1)
- Name the stages of anthropogenesis (10.2.6.6)

Classification

There is a vast variety of living things on our planet. There is no way of counting, never mind studying, every single individual organism alive today.

Organisms are classified according to similarities in structure, function and development. These similarities exist because organisms are related to each other, having arisen from common ancestors by evolution. The basic unit of classification is the species.



9.1 A horse and a mule: a mule is the offspring of a male donkey and a female horse. Horses and donkeys are different species as their offspring, the mule, is not able to reproduce

Benefits of classification

- To simplify the study of organisms (it is far simpler to study the features of a group such as flowering plants, than to learn the details of every type of flowering plant, about 250 000 in total).
- To allow scientists to communicate with each other.



9.2 A liger: the sterile offspring of a male lion and a female tiger

Classification means placing objects into groups based on similar characteristics.

Taxonomy is the science of classifying organisms.

Species

All domestic (tame) dogs are members of the same species. All such dogs can interbreed with each other to produce offspring, which themselves can reproduce.

Domestic cats are members of a different species from dogs. Dogs and cats cannot naturally interbreed with each other. For the same reason, oak and ash trees are different species.

A species is a group of similar organisms that are capable of naturally interbreeding with each other to produce fertile offspring.



So far, about 1.5 million species have been identified on Earth. Interestingly, insects alone make up about 750 000, or half, of all known species. Thousands of previously unidentified species are discovered every year. It is estimated that the total number of all species on Earth ranges between 5 and 100 million.

Variation within species

All living humans are members of the same species. Despite their many similarities humans may differ in features such as hair colour, skin colour and height. In the same way, individual plants of the same species (such as roses) may differ in traits such as flower colour, leaf shape or size of thorns.

These differences mean that each individual organism (such as a person) is unique. The individual variations do not hinder the ability of the organisms to interbreed successfully.

There are two types of variation: acquired and inherited.

Variation within a species means that in a group of successfully interbreeding organisms the individual members show different characteristics.

Acquired variation

Acquired variations are not inherited but are learned or developed during life.

Acquired variations are not genetically controlled. They are differences that form during life. Examples include the ability to walk, speak a language, ride a bicycle or use a computer.

Inherited variation

Examples of inherited variations are blood groups, eye/ear/hair colour, attached or detached earlobes, freckles and ear shape.

Inherited variations are controlled by genes.

Causes of inherited variations

Inherited variations arise as a result of sexual reproduction and mutations.

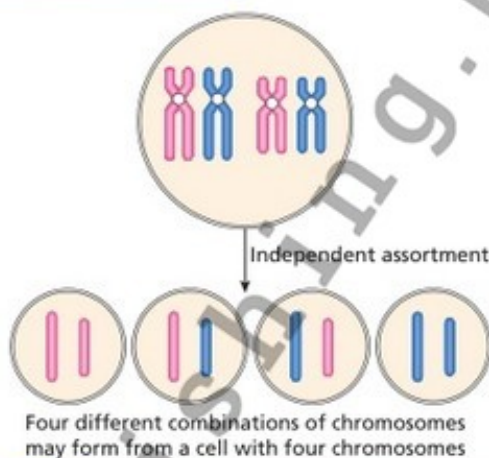


9.3 A father and son: tongue rolling is genetically inherited

Sexual reproduction as a cause of variation

Sexual reproduction is responsible for most of the variations that arise in each generation of offspring. Sexual reproduction causes genetic variation for three reasons.

- 1 Variations arise because of the independent assortment of chromosomes during meiosis. Human cells have 46 chromosomes. This means there are about 8 million different combinations of chromosomes available as a result of meiosis.
- 2 During meiosis, a process called crossing over takes place (details of crossing over are not required). This allows genes to be exchanged between chromosomes. This produces chromosomes that are a combination of the genes the mother and father inherited from their parents.
- 3 Finally, at fertilisation, owing to the random assortment of chromosomes in the egg and in the sperm, a vast range of variation is possible when a sperm and egg join.



9.4 Variation due to independent assortment in a cell with only four chromosomes

Mutations

Mutations can arise anywhere at random on a chromosome. This means that any gene, or group of genes, can be affected by mutations. However, cells contain enzymes that have a great ability to repair damage to DNA. This means that the number of mutations that survive is very low.

If a gene is altered it is very likely that the change in its sequence of bases will mean that the correct protein is no longer formed. The new version of a gene formed in this way by mutation is called a recessive allele.

Many mutations produce no change in the characteristics of a diploid organism. This is because the dominant allele on the second homologous chromosome can still produce the original protein.

A mutation is a spontaneous (or sudden) change in the amount or structure of DNA.

Harmful mutations

However, many mutations are harmful. Mutations in somatic (non-reproductive) cells may not be harmful. This is because the gene that is altered may not be active in the particular body cell affected. For example, if a skin cell suffers a mutation in a gene for saliva production, the cell will not suffer because saliva is not produced by skin cells.

Some somatic mutations are harmful. If the mutation causes an increase in the rate of mitosis, then a tumour may result.

Mutations in a gamete are often very serious. This is because the mutation may be inherited by the zygote and passed on to *all* the cells in the developing child. This may give rise to genetic defects in the child or even in the following generation.

Beneficial mutations

A very small number of mutations may be beneficial in that they produce an even better protein than the original one. These mutations are a source of evolution.

Causes of mutations

Mutations may arise naturally when DNA fails to produce exact copies of itself or when it fails to repair properly. Mutations such as these are called **spontaneous mutations**.

Mutagens are agents that cause mutations.

If mutagens are present, the spontaneous rate of mutation is speeded up. A mutagen that causes cancer is called a carcinogen. The main categories of mutagens are:

- Ionizing radiation such as x-rays, gamma rays, cosmic rays and ultraviolet (UV) radiation
- Chemicals such as formaldehyde, tobacco smoke, dioxins, caffeine and many drugs, preservatives and pesticides
- Some viruses.



To protect against mutation when being x-rayed, a heavy lead shield is used to absorb stray x-rays.

A high sun protection factor cream should be used when sunbathing. This may reduce the risk of developing skin cancer in later life as a result of exposure to UV rays from the Sun.

Tobacco smoke contains about 400 harmful substances, which are responsible for 90% of lung cancer deaths (they also cause many other diseases). Even passive smokers have a 35% increased risk of developing lung cancer.

9.5 A patient wearing a lead apron for protection against x-rays



Types of mutation

Gene (or point) mutations

Very often gene mutations are caused by changes in a single pair of bases. The altered gene is called an allele. Examples of gene mutations are:

- Cystic fibrosis (the inability to remove mucus from the lungs)
- Haemophilia (the inability to form proper blood clots)
- Albinism (lack of the skin pigment melanin)
- Cancers
- Sickle-cell anaemia.

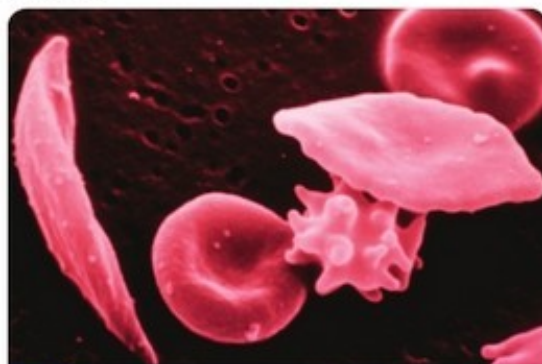
A gene (or point) mutation is a change in a single gene.

Sickle-cell anaemia as an example of a gene mutation

Sickle-cell anaemia is an inherited blood disorder caused by a mutation in the haemoglobin gene. The mutated gene forms a recessive allele.

A single copy of this mutation is found in about 10% of black Africans (who are healthy), but up to 1% have a double copy (i.e. they are homozygous recessive) and suffer from the disorder. It is also fairly common in people born near the Mediterranean.

A person with two copies of the recessive allele produces haemoglobin with one incorrect amino acid. This results in an insoluble form of haemoglobin that causes the red blood cells to take on a curved or sickle shape. This causes the breakdown and clumping of red blood, which in turn leads to paleness, weakness, heart failure, severe pains, damage to the brain and other organs and, very often, death.



9.6 Sickle-cell anaemia: note the mis-shaped red blood cells

Apart from treating the symptoms of the disorder, the most common treatment involves total blood transfusions. Such a treatment is only temporary and is not easily available in many parts of Africa. There is some hope that bone marrow transplants may someday provide a permanent cure for sickle-cell anaemia.

Chromosome mutations

Humans normally have 46 chromosomes in each somatic (non-reproductive) cell. If they gain a chromosome they will have 47, and if they lose a chromosome they will have 45 (i.e. $2n = 47$ or $2n = 45$).

Example of a chromosome mutation

Down syndrome (formerly called Down's syndrome) is an example of a chromosome mutation caused by the presence of one extra chromosome. The syndrome is usually caused by three number 21 chromosomes so that $2n = 47$ (whereas usually a person has only two number 21 chromosomes).

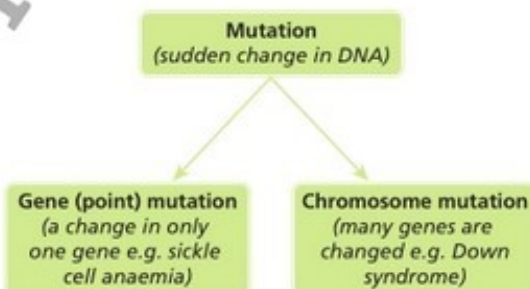
This disorder often arises from a fault in meiosis where the egg has two number 21 chromosomes (instead of one), with the sperm adding one more. The presence of the single extra chromosome produces a range of physical and mental features associated with Down syndrome.

Down syndrome is more common in children born to older mothers. The exact reason for this is not yet known.



9.7 Man with Down syndrome

A chromosome mutation is a large change in the structure or number of one or more chromosomes.



9.8 Summary of mutations

Evolution

Up until the early 1800s many people believed that species were fixed and unchanging. Since the start of the 19th century a number of theories of evolution have been suggested.

The most widely accepted modern theory of the mechanisms by which evolution takes place is based on the work of **Charles Darwin**.

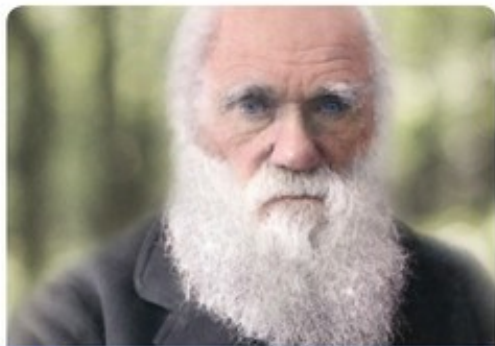
Darwin initially studied theology but later became a naturalist. He formulated many of his ideas aboard a research ship, the HMS *Beagle*. Much of his work was done in the Galapagos Islands, which are located in the Pacific Ocean, west of South America.

Darwin's theory was first presented in 1858, largely due to pressure from another naturalist, **Alfred Russel Wallace**, who had come up with the same ideas as Darwin while living in Borneo.

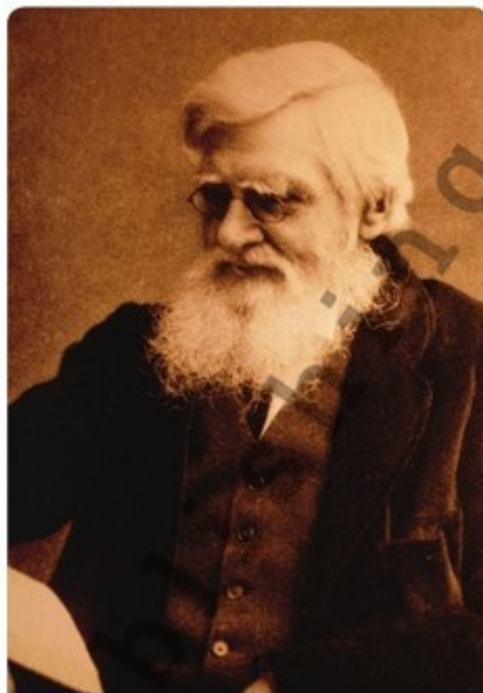
In 1859 Darwin published these ideas in his book *On the Origin of Species by Means of Natural Selection*. Since then this theory has been known as 'natural selection'.

Evolution is the way in which living things change genetically to produce new forms of life over long periods of time.

Although almost everyone accepts that evolution takes place (i.e. that living things have changed over many millions of years), not everyone agrees that these changes take place by natural selection. For instance, many people believe that the changes were due to some form of divine intervention.



9.9 Charles Darwin (1809–1882)



9.10 Alfred Russel Wallace (1823–1913)

Theory of natural selection

The theory of evolution by means of natural selection is based on three observations and two conclusions derived from these observations. These ideas are outlined below.

Observation 1: overbreeding

Darwin noted that organisms produce large numbers of offspring. For example, trees produce thousands of seeds and oysters lay millions of eggs.

Observation 2: population numbers remain constant

The number of organisms of the same species (called a population) in an area will continue to increase until the environment can no longer support any more. Then the number of organisms stays more or less the same.

Conclusion 1: there is a struggle for existence

If more offspring are formed and the environment cannot support all of them, then there must be competition for scarce resources. This means that animals compete for food, water, shelter and mates. Plants compete for space, light, water and minerals.

Observation 3: inherited variations occur in populations

The members of a population or species show genetic or inherited differences. These variations may arise from sexual reproduction, mutations or genetic engineering (variations arising due to the environment are not passed on, e.g. broken legs, learned abilities).

Conclusion 2: natural selection

Those organisms that have variations that enable them to adapt better to their environment will survive and reproduce. They will pass their variations on to the next generation. Organisms with unfavourable variations will not survive and they will not be able to pass on their variations to the next generation.

Natural selection is also called survival of the fittest, but this expression is misleading. Natural selection is not about fitness in terms of physical or mental abilities. Instead, it relates to the suitability of a species (or organism) to its environment. The essence of natural selection is how well adapted organisms are to their environment.

Natural selection is the process by which those organisms with genetically controlled characteristics that allow them to be well adapted to their environments will survive and reproduce to pass on their genes to following generations.

Speciation

If organisms are created that can obtain more food, resist disease or produce more offspring, then they will be 'selected' by nature. This means they may live longer and reproduce more often and so be allowed to pass on their genes.

In time, the accumulation of slight changes results in the formation of organisms that can no longer interbreed. A new species is said to have formed, and speciation is said to have occurred. The formation of new species does not mean that evolution ceases to take place. Evolution is a continual process and is taking place in all species (including humans) at present.

Speciation is the production of new species as a result of evolution.

Evidence for evolution

There are a range of different scientific approaches to studying evidence of evolution. These include the study of fossils, comparative anatomy, comparative biochemistry, embryology and geographic distribution.

The study of fossils

A **fossil** is the remains of something that lived a long time ago (or some indication of something that lived a long time ago).

One of the best sources of evidence for evolution is **palaeontology** (the study of fossils).

Examples of fossils include entire organisms, shells, bones, teeth, seeds, pollen grains, leaf prints, footprints and even the remains of faeces.

Fossil evidence for evolution

The information outlined below indicates that organisms have changed over time. It does not prove that these changes were due to natural selection.

- **Fossils can be aged.** This can be carried out by reference to the depth at which they are found in a rock or soil formation, or by measuring the amount of radioactive decay. This allows fossils to be compared according to a timescale.



9.11 A 30-million-year-old fossil fly in amber (solidified tree resin)



9.12 Dinosaur footprint: 120 million years old

- **Fossils show changes when compared with modern organisms.** These changes can be related to the time difference between when the fossilised organisms existed and the present.
 - ▶ Some fossilised organisms no longer occur as living organisms (they are extinct, e.g. the dodo and dinosaurs).
 - ▶ In other cases, there is no fossil record of modern species. This could be due to the modern organism being recently formed and therefore having no fossil record.
- **The more modern fossils show increased complexity.**
- **Very often the fossil evidence can be linked to environmental change,** i.e. organisms had new environments to which they had to adapt. For example, 65 million years ago dinosaurs and many plants became extinct. At this time a layer of dust (rock) containing the element iridium was laid down. Iridium is rare on Earth but common in meteorites.

This suggests there may have been a huge meteorite impact, creating large amounts of dust. The dust is thought to have reduced the amount of heat and light entering the Earth's atmosphere from the Sun. This may have resulted in the sudden, mass extinction of plants and animals.

Evolution of the horse

The fossil record of the modern horse is very well documented. It covers a time span of about 60 million years and involves many hundreds of species, most of which are now extinct.

There are many trends to be seen in the evolution of modern horses. The following account deals with one change: the height of the animal.

- The ancestor of the modern horse developed about 60 million years ago. These animals were about the size of a fox (0.4 m high).
- Fossils from about 30 million years ago show that the ancestors of the horse were larger (about the size of a German shepherd, 0.6 m high).
- Fossils from 15 million years ago show the existence of creatures that were the size of a Great Dane (1 m high).
- The modern horse first evolved about 1 million years ago. Modern horses are normally about 1.6 m high.

Timescale	Height
1 million years ago to the present	 1.6 m
15 million years ago	 1.0 m
30 million years ago	 0.6 m
60 million years ago	 0.4 m

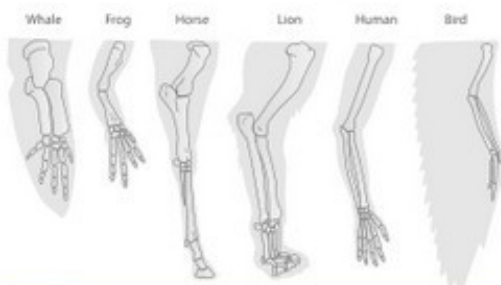
9.13 Evolution of the modern horse



9.14 Artist's impression of the first horse, 60 million years ago

Comparative Anatomy

The study of comparative anatomy, of great interest to Darwin, is a further means of providing evidence for evolution. Darwin studied the forelimbs of a variety of animals, including humans, birds and bats and noted strong similarities (see figure 9.15). These commonalities, in his view, pointed to similar origins and he used this evidence to point to a common ancestor for modern forms. Darwin posited that any differences in structure or function are the result of adaptations to the special needs of modern organisms. He also observed that animals have structures that they do not use – possibly due to environmental changes. These structures often degenerate and become smaller in size compared with similar organs in other organisms. In humans, these vestigial organs include the appendix, which may have been used in digestion; the fused tail vertebrae; the wisdom teeth and the muscles controlling movement of the ears and nose.



9.15 Similarities in forelimbs of humans and five other animals despite the difference in functional use.

Research the various stages in the development of the human species. Name each stage and prepare a slide with visuals to present to the class.

Comparative Biochemistry

Studies in modern biochemistry indicate that there is a biochemical similarity in all living organisms. Mechanisms for trapping and transforming energy and for building proteins from amino acids are strikingly similar. All living organisms share an almost identical genetic code with DNA and RNA as the mechanisms for inheritance and gene activity. Such uniformity in biochemical organisation accounts for the diversity of living organisms and points to evolutionary relationships.

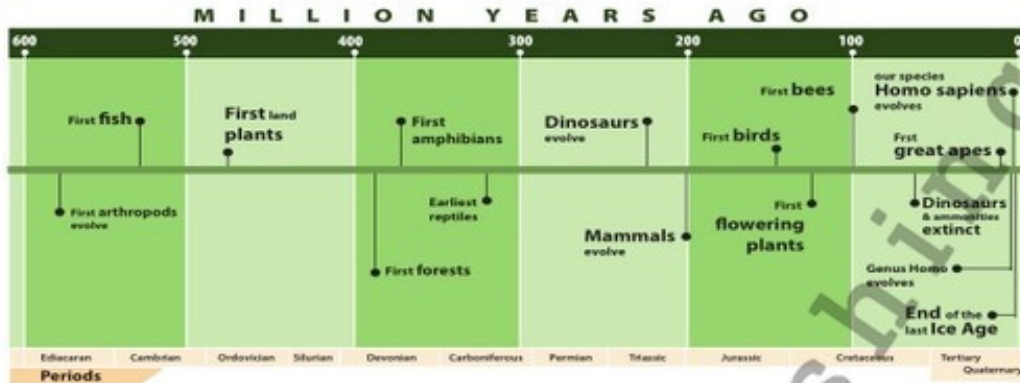
Embryology

Darwin studied embryos and their development and used his findings to support the theory of evolution. He noted many similarities among embryos of complex animals such as humans, chickens, frogs, reptiles and fish and suggested that many embryonic stages are inherited from a common ancestor. Darwin observed that early human embryos are similar to those of birds, fish and rabbits: all have gill slits, a two-chambered heart and a tail with muscles to move it. As the embryo grows and develops, these similarities disappear.

Geographic distribution

Isolated environments, such as the islands of New Zealand and Australia, have animal populations which exist nowhere else in the world. Darwin famously studied life forms on the Galapagos islands and discovered unique species. He noted 13 different species of finch found only on the island, which he believed had evolved from a common ancestral group. This geographic distribution of species can be considered as further evidence for evolution.

Timeline of life on earth

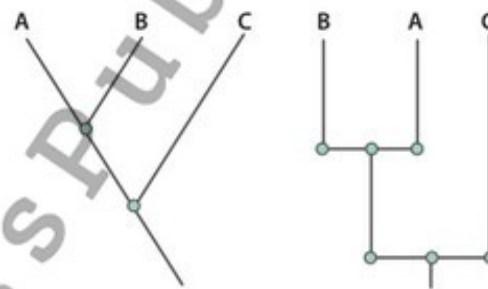


9.16 A timeline of the last 600 million years, showing major events in evolution.

Cladograms

Cladograms are a form of tree diagram used to show the similarities and differences between different species. The key feature on a cladogram is the branching points that are called nodes. A node marks the point of a speciation event, the point at which a common ancestor splits into two or more species. The term clade refers to a group of species which evolved from a common ancestor and share characteristics.

Common diagrammatic forms used for cladograms are shown in figure 9.17



9.17 Cladograms

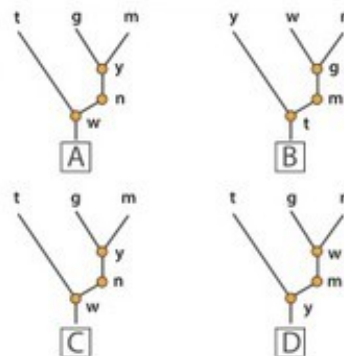
Look at the figures [a-d] below in figure 9.18 and say which best matches the information here:

G, M, N and T are all related species of organisms.

Species W is an extinct recent common ancestor of species G and N. W, G and N all evolved from species M.

Species T is the least related to the others and extinct. Species Y is its most recent phylogenetic link to the other species.

Techniques involving evidence from amino acid sequences of certain proteins and base sequences of genes are being used to further phylogeny studies and leading in some cases to a reclassification of the evolutionary origins of some species. A recent example of such reclassification is the Figwort plant family (Scrophlahulariaceae)



9.18 Cladograms

The Molecule Clock

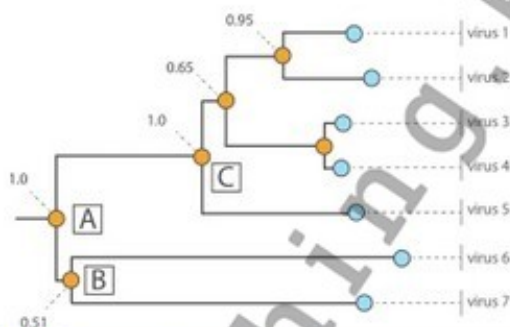
The molecule clock is the term used to refer a calculation of the number of mutations, which accumulate in the gene sequences of different species over time. Establishing mutation rates through the sequencing of nuclear DNA and mitochondrial DNA, allows scientists to work out the point in prehistory when two or more life forms diverged.

Phylogenetic Trees

Another form of diagram that can represent information relating to speciation events is the phylogenetic tree. A basic phylogenetic tree is shown in figure



Research the differences between cladograms and phylogenetic trees in terms of the detailed information they represent. In groups produce a slide presentation to present this with examples of each form of diagram.



Questions on Module 9

- Explain what is meant by:
 - Classification
 - Taxonomy.
- What is a species?
 - Name two animal and two plant species.
 - Why are lions and tigers considered to be different species?
- The members of a species show genetic variations.
Name three genetic variations that are visible in (a) Humans (b) A named plant.
- Why are all humans considered to be in the same species?
- What is meant by biological variation?
 - Distinguish, giving two examples in each case, between acquired and inherited variation.
- Name the cause of inherited variation.
- What is a mutation?
 - Explain why many mutations may not be serious.
 - What are mutagens?
 - Name two types of mutagen.
 - Why should sun creams with a high protection factor be used during sunbathing?
- Ozone is a gas that absorbs ultraviolet radiation in the atmosphere.
Why does a reduction in the amount of ozone in the atmosphere represent a threat to human life?
- Distinguish between gene and chromosomal mutations.
 - Give one example from each of the categories of mutations named in part (a) and give details of the effects of the named condition.
- What is evolution?
 - Name the two scientists who first proposed the theory of evolution by natural selection.
- A population of rabbits living on an island experience a struggle for existence.
 - What two observations did Darwin make to suggest a reason for such a struggle?
 - Suggest four resources for which the rabbits might struggle.
- What is a fossil?
 - Give four examples of different types of fossil that have been discovered.
 - State two methods used to date fossils.
- Explain three ways in which the theory of evolution is supported by the study of fossils.
- Outline any way in which fossils show how the modern horse has evolved.
- State the significance of each of the following in the theory of evolution by natural selection.
 - Organisms produce more offspring than their environment can support.
 - Organisms show genetically controlled variations.
 - A variation that does not improve the organism's ability to reproduce is of no value in terms of evolution.
 - Organisms that reproduce asexually (using only mitosis) tend to evolve more slowly.

Applied genetics

Applied genetics involves the manipulation of hereditary characteristics in order to improve or produce desirable characteristics in offspring. Desirable characteristics might include improved fertility, increased milk production or better growth rates in breeds of cattle. For centuries, farmers have used controlled breeding (also known as selective breeding) in plants and animals to concentrate desirable traits in offspring by careful selection of parents with those traits. However, with the advent of artificial insemination and embryo transplantation the number of offspring from genetically superior parents can be increased far beyond the numbers that would be possible by natural means. In addition, genetic engineering allows the production of new plant species which contain genes from unrelated organisms, giving us a new generation of plants known as genetically modified organisms.

Advent:
arrival or development.

Selective breeding

Selective breeding is also known as artificial selection and it can be used to eliminate undesirable traits. Selective breeding of animals produces breeds and selective breeding of plants produces varieties or cultivars. The selective breeding of plants and animals can be divided into inbreeding and crossbreeding.

Selective breeding is the process of breeding animals or plants with desirable traits and concentrating those desirable traits in their offspring.

Inbreeding

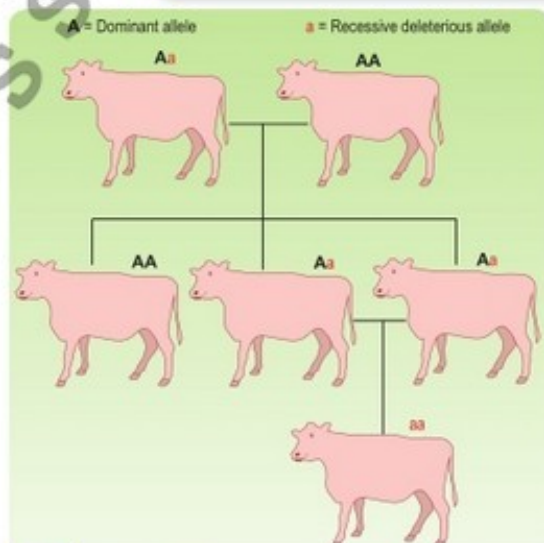
Inbreeding has the advantages of fixing desirable genetic traits such as high milk yields in purebred dairy cattle (e.g. Holsteins) and creating uniformity among the offspring. Inbreeding produces pedigree animals (purebred animals) whose ancestry (lineage) is known and the animal can then be registered with a breed society.

The disadvantage of inbreeding is that it also concentrates undesirable traits. Most animals carry undesirable traits. However, the genes for these traits tend to be recessive and are hidden by the presence of the dominant, normal gene. Closely related animals have similar genes and inbreeding increases the inheritance of similar genes, leading to homozygous genotypes (e.g. aa) as shown in Fig. 18.1. These undesirable recessive genes are often termed deleterious, since they can be harmful or unwanted.

In cattle, mulefoot is a recessive trait that is caused by the inheritance of two recessive genes, one from each parent. This causes the two toes on the front feet of the calf to be fused together. The offspring that inherit mulefoot are homozygous (bb) for the trait.

Some cases of homozygous recessive genes can be fatal, leading to the failed conception in a cow. In these circumstances, the recessive gene is called a lethal gene, since it causes the death of the organism.

Inbreeding is the mating of closely related animals, which increases the chances of offspring being affected by undesirable recessive traits.



9.20 Inbreeding in cattle

Deleterious: causing harm or damage.
Undesirable: not wanted.
Lethal: causes death or is life-threatening.

Lethal genes are self-limiting: organisms that inherit two copies of the gene cannot pass them onto their offspring. Most animal species carry some recessive lethal genes, which have no effect on the organism when they are present as a heterozygous genotype (Bb).

A study conducted into the levels of inbreeding in Irish dairy and beef herds concluded that while the levels of inbreeding were low, they were slowly rising annually in Hereford, Simmental and Holstein-Friesian herds. The study also documented that inbreeding resulted in inbreeding depression, which refers to a reduction in the performance of an animal as a result of inbreeding.

The study found that low levels of inbreeding in Holstein-Friesians led to a reduction in milk, fat and protein yields and there was an increase in their somatic cell count. It was observed that inbreeding had its greatest effects on the fertility of the Holstein-Friesians, leading to a greater chance of calving difficulties and increased calving interval.

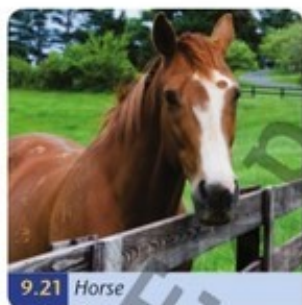
Inbreeding depression is the reduction in fertility, production and health in a given population as a result of inbreeding.

Crossbreeding

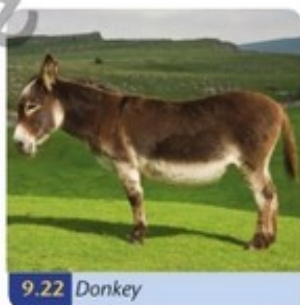
Crossbreeding or outbreeding involves the mating of animals or plants from two different breeds, varieties or species. The offspring in many instances inherit favourable genes from both parents, leading to improved health traits over either parent. This is referred to as hybrid vigour or heterosis.

Crossbreeding involves the mating of animals or plants from two different breeds, varieties or species. **Hybrid vigour (heterosis)** is the increased productivity displayed by offspring from genetically different parents.

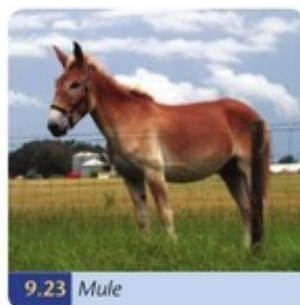
Crossbreeding is the opposite of inbreeding: it reduces the risk of harmful recessive genes being displayed in the phenotype by increasing the number of heterozygous pairs of genes. In its extreme, it can involve the crossing of two different species. This is the case when a female horse is crossed with a male donkey and the resulting offspring is called a mule. Mules demonstrate hybrid vigour: they have greater strength, are hardier animals with greater disease resistance and have a longer lifespan than their parents. The only drawback is that mules tend to be infertile and unable to reproduce.



9.21 Horse



9.22 Donkey



9.23 Mule

Countries such as New Zealand, Canada and the United States have conducted much research into the benefits of crossbreeding in dairy herds. Successive generations of inbreeding among purebred dairy cows (e.g. Holsteins) had led to decreased fertility rates and decreased productivity over the lifespan of the cow. In addition, the dairy industry was changing with farmers' milk payments increased by high milk solids content (protein and butterfat) and decreased for high water content of milk.

Purebred Holstein cows are the highest milk producers; however, milk protein and fat is low in comparison to volume of milk produced. Jersey cows, on the other hand, have the highest milk solids in their milk, but low milk yields. When Holsteins are crossed with Jerseys, the offspring have increased milk production from the Holstein and increased milk solids (protein and butterfat) from the Jersey. In addition, the offspring have increased fertility, health and lifespan in the dairy herd, which reduces the replacement rate.

Uniformity: consistency, regularity or evenness.

Crossbreeding also comes with disadvantages. There is a loss of hybrid vigour with subsequent crossing of the hybrids. This results in a reduction in the uniformity of the phenotype in the offspring. Holstein-Jersey cross males have very poor conformation and, as a result, are not suitable for beef production. In addition, crossbreeding requires the maintenance of two purebreds in order to produce the crossbreds.



9.24 Holstein cow



9.25 Jersey cow



9.26 Holstein-Jersey cross

Crossbreeding in F1 hybrid seed varieties

Crossbreeding is extensively used for the production of F1 hybrid seed varieties. These hybrid crosses are often stronger, have greater disease resistance and higher yields than their purebred parents. These F1 hybrid seeds are derived from the crossing of two genetically different parent cultivars. For simplicity, we will refer to these parent plants as cultivar A and cultivar B. These parent plants have been inbred by repeated self-pollination over several generations so that they are practically homozygous for their traits. These parent plants are referred to as breeding stock. These two different cultivars 'A and B' and then crossed by hand pollination. This involves the removal of the immature anthers that produce the pollen from cultivar A, and the dusting of cultivar A's stigma with the male pollen from cultivar B. This then produces the seeds, which are the hybrid AB. The F1 hybrids benefit from hybrid vigour and the hybrids are uniform in phenotype.

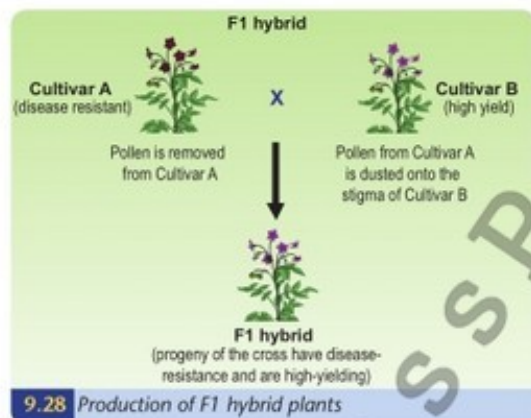
➡ **Cultivar:** a variety of plant that has been produced by selective breeding.

➡ **Variability:** variety, alteration or inconsistency.

The production of F1 hybrid seeds is an expensive process, since the parent plants 'cultivars A and B' must be crossed every year. If the F1 hybrids are crossed for an F2 generation, the offspring have greater variability compared to the F1 and, as a result, lose some of the hybrid vigour of the F1.



9.27 F1 seed variety packets



9.28 Production of F1 hybrid plants



9.29 A hybrid plant



Commercial potatoes are produced asexually using techniques such as micropropagation to produce many seed potatoes. This ensures that the potatoes are uniform in their genetic make-up and thus their phenotype. However, these seed potatoes are not true seeds. In order to produce true seeds, the potato plant must reproduce sexually by the transfer of pollen from one potato plant to the stigma in a flower of another potato flower. A fruit (similar in appearance to a tomato) develops where the flower was. This is the true fruit of the potato plant and inside this fruit are the true potato seeds. Using controlled crosses, breeders will produce true potato seeds when developing new varieties of potatoes.

Questions on Module 9

- 16 Explain the term applied genetics.
- 17 Outline the advantages and disadvantages of inbreeding.
- 18 Crossbreeding between Holstein and Jersey is becoming common in the dairy herd. Outline the advantages and disadvantages of crossbreeding Holsteins with Jerseys.
- 19 Explain the term hybrid vigour.
- 20 Describe the production of F1 hybrid seed varieties. What are the advantages and the disadvantages of the production of F1 hybrid seed varieties?
- 21 Distinguish between each of the following pairs of terms:
 - (a) Inbreeding and crossbreeding
 - (b) Selective breeding and cloning

Module 10 The nervous system

Learning objectives

- Describe and explain the initiation and transmission of the action potential in myelinated neuronal axons [10.1.7.1](#)
- Explain the value of the refractory period and the myelin sheath [10.1.7.2](#)
- Study the structure and functions of the spinal cord and brain [10.1.7.3](#)
- Describe how mechanoreceptors react to changing the stimulus (Pacini corpuscles) [10.1.7.4](#)
- Establish the relationship between the structure and function of the cholinergic synapse [10.1.7.5](#)

Introduction

In animals, the nervous system and the endocrine system are responsible for the coordination of activities in the body. They allow the animal to respond to internal and external changes called stimuli.

The nervous system is specially adapted for the rapid responses that an animal makes. It is divided into:

- The central nervous system (CNS), which consists of the brain and spinal cord
- The peripheral nervous system (PNS), which consists of a vast network of nerves (in the form of nerve fibres) that carry messages between the CNS and the rest of the body.

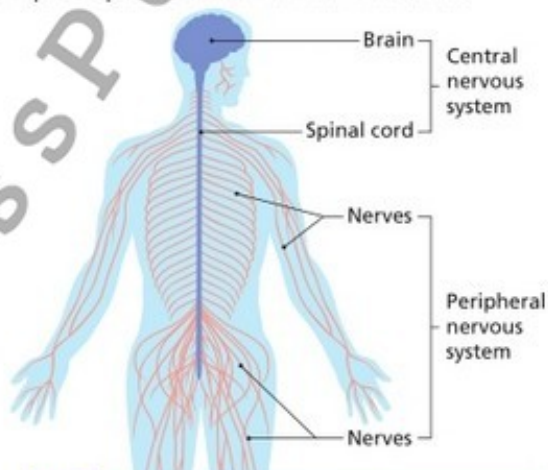
In order to carry out the correct response to a stimulus, four processes are involved.

- **Reception.** The stimulus must be detected. This is the function of receptors, neurons (nerve cells) and sense organs.
- **Transmission.** The message passes along the neurons. The neurons in the PNS carry messages from receptors to the CNS and from the CNS to effectors, such as muscles or glands.
- **Integration.** The incoming messages are sorted and processed and a response decided upon. This occurs in the CNS, especially in the brain.
- **Response.** This is carried out by the effectors (i.e. muscles or glands) when they are stimulated by neurons.

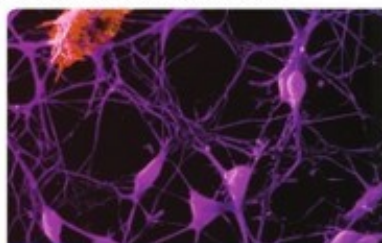
Neurons

Neurons are the basic units of the nervous system and are specialised to carry information (as electrical impulses) from one place to another.

There are three types of neuron: sensory, motor and interneurons. Not only do neurons vary in type,



10.1 The parts of the nervous system



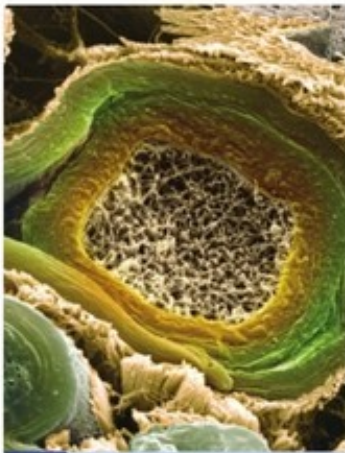
10.2 A network of neurons: cell bodies and dendrites are visible

they also differ in size. Neurons in the brain are very tiny, whereas neurons connecting the spine and the feet may be over 1 metre long.

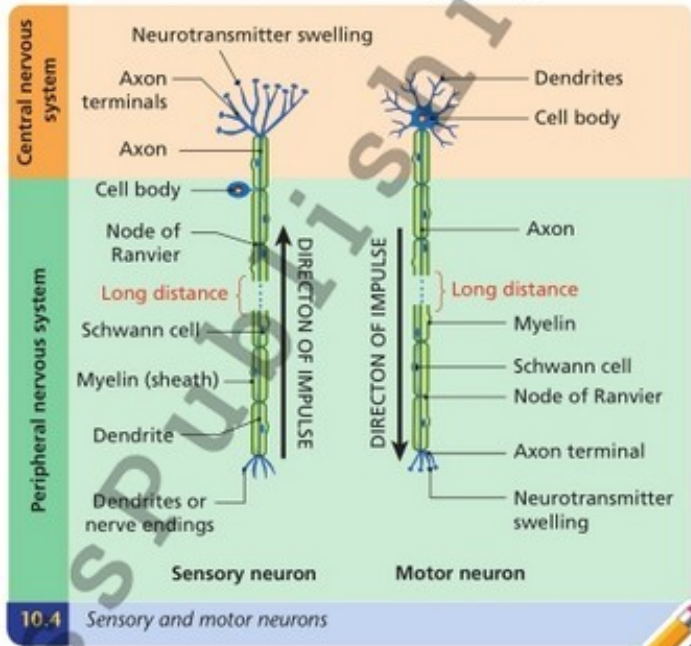
Structure of neurons

The typical structure of a sensory and a motor neuron is shown in diagram 10.4.

A **neuron** (or neurone) is a nerve cell. A **sensory** (or afferent) neuron takes a message from a sense organ to the CNS. A **motor** (or efferent) neuron takes a message from the CNS to a muscle or a gland.



10.3 Section of a neuron: the axon (beige) in the centre is surrounded by myelin (orange/green), which in turn is enclosed by a Schwann cell (green)



10.4 Sensory and motor neurons

Functions of the parts of neurons

- A **receptor** is a cell or group of cells that detects a stimulus.
- **Nerve endings** connect sensory neurons to receptor cells or sense organs.
- **Dendrites** are fibres (often highly branched) that carry impulses **towards** the cell body.
- **Axons** carry impulses **away from** cell bodies.
- **Schwann cells** produce the myelin sheath.
- The **myelin sheath** is a fat-rich layer that insulates the electrical impulses.
- The **cell body** contains a nucleus and cell organelles. In particular cell bodies contain mitochondria, which provide energy for the movement of nerve impulses. Each cell body forms the dendrites and axons that may emerge from it as well as neurotransmitter chemicals. The cell bodies of sensory neurons are located outside the CNS. The cell bodies of motor neurons are located within the CNS.

Axons start with the letter 'a' and carry impulses away from the cell body.

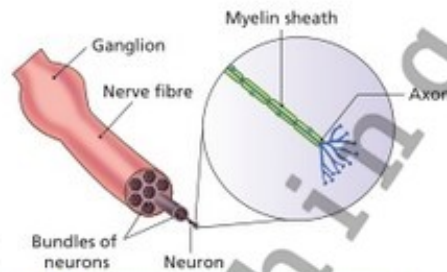
A **ganglion** (plural: ganglia) is a group of cell bodies located outside the CNS. An **interneuron** (also called an intermediate, relay or association neuron) carries information between sensory and motor neurons.

? In multiple sclerosis (MS), patches of myelin degenerate in the CNS. As a result, the passage of nerve impulses is impeded and the person suffers symptoms ranging from numbness and tingling to difficulty in moving or paralysis and loss of bladder control.

- **Axon terminals** are branches formed by the splitting of an axon. Each of these small branches carries an impulse to a swelling called a neurotransmitter swelling.
- **Neurotransmitter swellings** release chemicals that carry the impulse from one nerve cell to another. The chemicals are called neurotransmitters. Neurotransmitters are stored in vesicles in the swellings.

Nerve fibres

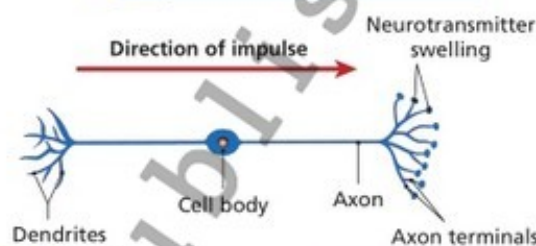
Many axons or dendrites often combine to form nerve fibres (sometimes simply called a nerve). Axons can be very long, e.g. those that run from the spine to the feet may be over 1 metre in length.



10.5 Relationship between a nerve fibre and neurons

Interneurons

Interneurons are short neurons found between motor and sensory neurons in the CNS. They are not enclosed in myelin sheaths.



10.6 Structure of an interneuron

Transmission of nerve impulses

A stimulus of sufficient strength arriving at a neuron causes an electrical current or impulse to travel along the dendrite and axon to the neurotransmitter swellings. The movement of the electrical impulse along a neuron involves the movement of ions (charged particles) in and out of neurons. This movement uses energy and is an active process.

Features of nerve impulses

Resting neuron

A resting neuron is a neuron that is not carrying an impulse.

Threshold

A stimulus below the threshold has no effect, but one that is at or above the threshold causes an electrical impulse to travel along the axon. Some people are said to have a higher threshold for pain or for high temperatures. This means they can tolerate more pain or higher temperatures before their nervous system reacts.

The threshold is the minimum stimulus needed to cause an impulse to be carried in a neuron.

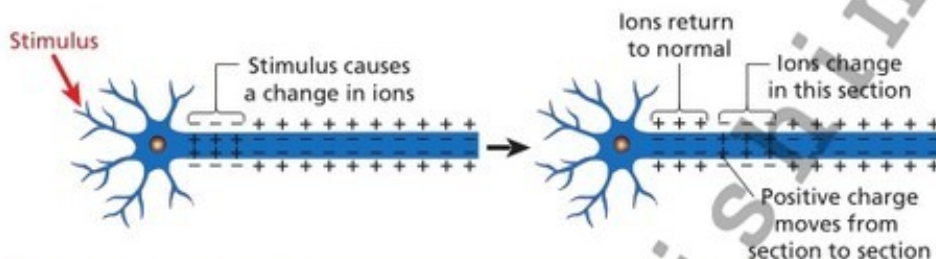
The 'all or nothing' law states that if the threshold is reached an impulse is carried, but if the threshold is not reached no impulse is carried.

'All or Nothing' law

The 'all or nothing' law means that an impulse is either carried or not carried. If the threshold is reached a message is sent. No matter how strong the stimulus is, once it is above the threshold level, the same impulse is carried. This means that a mild stimulus or a severe stimulus will cause the same impulse to be sent along any axon. Sensitivity to different degrees of stimulation (e.g. mild versus severe pain) depends on the number of neurons stimulated and the frequency with which they send their impulses.

Movement of impulse

Once the threshold is reached the axon (or dendrite) changes its permeability to ions. This allows for the transmission of an impulse. An impulse will cause a section of a neuron to change its permeability to ions. This will cause the next section to change its permeability and the impulse will 'jump' to that section. Once the impulse has moved along, the area behind is restored to the original state. This pattern of movement of ions continues along the entire length of the neuron.



10.7 Movement of an impulse in a neuron

Refractory period

There has to be a slight delay of a fraction of a second between any two impulses. This time span is necessary to allow the impulse to pass from one neuron to a second neuron (at a synapse, as explained later in this module). This tiny time span is called the refractory period.

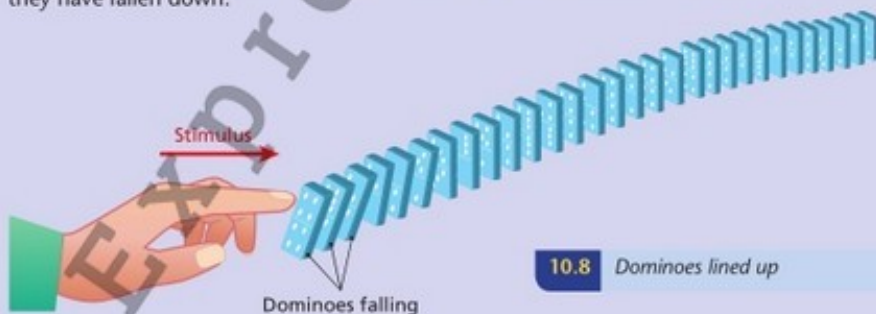
The refractory period is a short time span after a neuron has carried an impulse during which a stimulus fails to cause a response.



The transmission of a nerve impulse can be compared to a set of dominoes lined up, as shown in diagram 10.8.

- If the first domino is touched lightly the dominoes do not fall. This is the equivalent of the threshold not being reached, and so no impulse is carried.
- If the first domino is pushed hard enough, it and all the other dominoes will fall. The threshold was reached and an impulse was carried.

The refractory period can be compared to the time needed to stand the dominoes up again after they have fallen down.



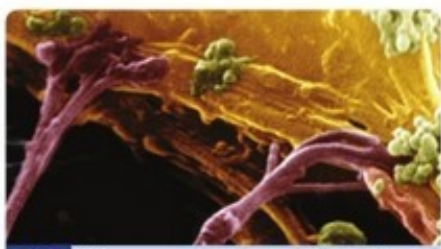
10.8 Dominoes lined up

Speed of impulse

The speed at which an electrical impulse travels along a neuron depends on whether myelin is present or absent around the neuron.

- When myelin is present the impulse 'jumps' from one node of Ranvier to the next. This means the impulse travels very fast.
- When myelin is absent the impulse must travel along the entire length of the neuron. This slows down the speed of the impulse.

The speed at which the electrical impulse travels is also dependent on the diameter of the dendrite or axon. The larger the diameter, the faster the impulse travels.



10.9 Synapses: axon terminals (purple) connecting to a cell body (yellow)

Synapse

Synapses are commonly found between the axon terminals of one neuron and the dendrites of another neuron. Normally the gap is as small as 0.00002 mm.

The number of synapses associated with each neuron is very large, ranging from 1000 for a cell body in the spinal cord up to 10 000 for cell bodies in the brain.

A synapse is a region where two neurons come into close contact. A **synaptic cleft** is the tiny gap between the two neurons at a synapse.

Passage of an impulse across a synapse

Activation of neurotransmitter

Electrical impulses cannot cross a synapse. Instead, the impulse stimulates the neurotransmitter swellings (at the end of the axon) in the pre-synaptic neuron to release a chemical substance that diffuses rapidly across the synaptic cleft. These chemicals are called **neurotransmitters**.

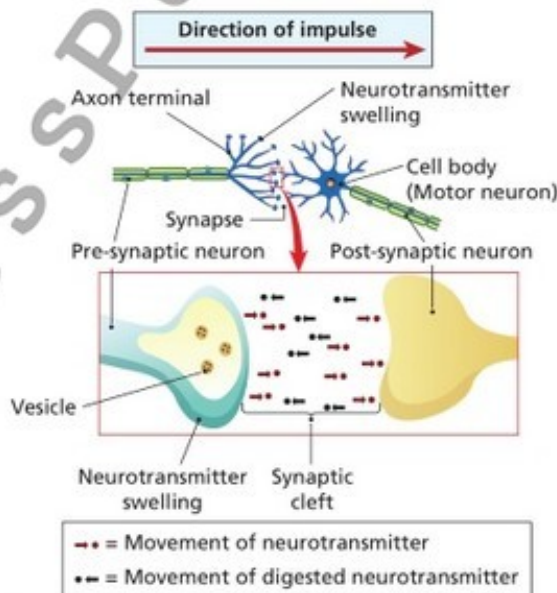
Some neurotransmitters are made in the cell bodies of the neurons, whereas others are formed in the neurotransmitter swellings. The enzymes needed to make these latter neurotransmitters are made in the cell body and transported to the swellings. Over 60 different neurotransmitters are known, the most common being **acetylcholine (ACh)**, **noradrenalin** (also called norepinephrine) and **dopamine**.

Inactivation of neurotransmitter

The neurotransmitter diffuses across the synaptic cleft. It then combines with receptors on the post-synaptic neuron and is broken down by enzymes. The digested neurotransmitters are reabsorbed back into the neurotransmitter swellings. This allows them to be recycled and reused upon the arrival of the next impulse. The breakdown of the neurotransmitters causes the electrical impulse to be regenerated and to be transmitted onwards.

Electrical impulse → Chemical impulse → Electrical impulse

10.11 The interchange that occurs at a synapse



10.10 Events at a synapse

Functions of synapses

- 1 They **transmit impulses** from one neuron to another neuron or to an effector (muscle or gland).
- 2 They **control the direction of the impulse**. This is due to neurotransmitter swellings being found only on the pre-synaptic side of the synapse. In this way they act as valves (i.e. they allow only a one-way flow).
- 3 They **prevent overstimulation** of effectors. This happens when the neurotransmitter ceases to be produced due to constant stimulation. The impulse is therefore inhibited and the

effector ceases to be stimulated.

- 4 This is why we tend to get used to stimuli such as pain and noise and are stimulated only by changes in these inputs.
- 5 The **impulse can be blocked** by certain chemicals (drugs). This is important in controlling pain and some psychiatric disorders.



10.12 The human brain

The central nervous system (CNS)

The brain

The senses and individual sensory neurons act as receptors for incoming stimuli. Electrical impulses are passed along the sensory neurons (dendrites and axons) to the CNS, and in particular to the brain. The brain acts as an interpreting centre to sort and process the incoming impulses and decide on a response.

The human brain is composed of about 85 000 million (85 billion) neurons. The cell bodies and synapses form the grey matter of the brain, with the nerve fibres (dendrites and axons) forming the white matter. The brain uses about 20% of the body's energy.

Both the brain and spinal cord are protected by bone (the cranium and vertebral column respectively) and are covered by three membranes called **meninges**. The space between the inner

two meninges is filled with cerebrospinal fluid. This fluid acts as a protective shock absorber and as an exchange medium between the blood and brain.



Meningitis is an inflammation of the meninges that enclose the brain and also the nerves in the spinal cord. Meningitis occurs more commonly in children than adults, but can occur at any age. There are two causes of meningitis: a virus and a bacterium.

- Viral meningitis is a more common and less severe infection. It causes irritability, headache and fever. In severe cases it results in neck ache. There is no specific treatment and the symptoms normally disappear within a week or two.
- Bacterial meningitis is much more dangerous. Along with the symptoms described above, it causes skin rash, vomiting, intolerance of bright light, inability to bend the neck down, convulsions and even coma and death. Bacterial meningitis is treated with antibiotics. Vaccines are available for some forms of bacterial meningitis.

Structure of the brain

Cerebrum

The cerebrum is the largest part of the brain. It contains about 75% of the neurons in the brain. It is divided into two halves: the right and left cerebral hemispheres.

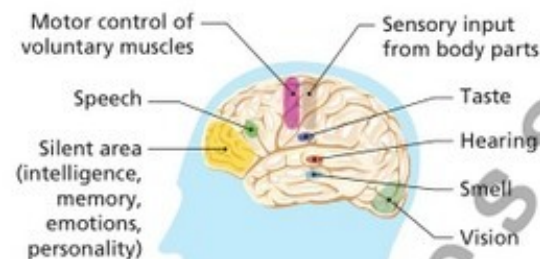
The functions of the cerebrum include:

- Controlling voluntary movements
- Receiving and interpreting impulses from the sense organs
- Thinking
- Intelligence
- Memory
- Language
- Emotions
- Judgement
- Personality.

The right hemisphere controls the left-hand side of the body. Neurons from the left hemisphere control the right-hand side of the body. This is why a stroke (a blood clot in the brain) may paralyse only one side of the victim.

Each hemisphere is specialised to function in different ways.

- In general, the left side is dominant for hand use (i.e. most people are right-handed), language, mathematics, analysis and logic.
- The right side specialises in art, music, shape recognition and emotional responses.



10.14 Functions of the cerebrum

The cerebellum controls:

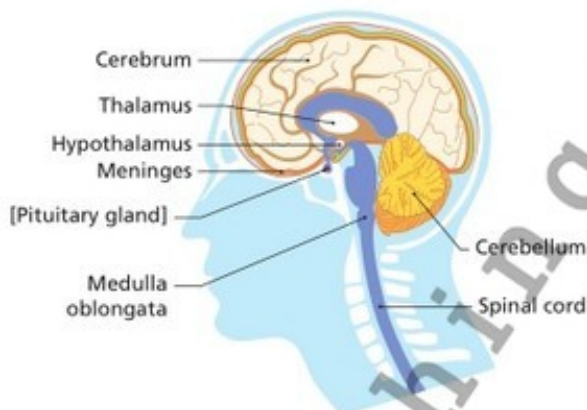
- Muscular coordination (and allows smooth, refined muscular action)
- Balance.

The responses of the cerebellum are **involuntary**, once the process has been learned. For example, walking is originally controlled by the cerebrum, but once we learn to walk the cerebellum takes over and walking is controlled involuntarily.

Medulla oblongata

The medulla oblongata connects the spinal cord with the rest of the brain. It controls involuntary actions such as:

- Breathing
- Swallowing
- Salivation
- Sneezing.
- Blood pressure
- Coughing
- Vomiting



10.13 Structure of the brain

The functions associated with different parts of the cerebrum are shown in diagram 10.14.

The outer part of the cerebrum, called the cerebral cortex, is grey. The inner, white matter of the cerebrum contains millions of nerve fibres. These connect different areas of the cortex and the two sides of the brain (which are connected by a structure called the corpus callosum).

Cerebellum

The cerebellum is the second largest section of the brain. It is heavily **folded**.

Thalamus and hypothalamus

The **thalamus** is located below the cerebrum.

- It acts as a sorting centre for the brain (by sending all incoming impulses to the correct part of the brain).

The **hypothalamus** lies below the thalamus. Its functions are to:

- Regulate the internal environment of the body (homeostasis) by monitoring body (blood) temperature, appetite, thirst, osmoregulation and blood pressure
- Link with the pituitary gland to regulate the production of many hormones.

The hypothalamus is thought to be the link between the mind (or brain) and the body.

Pituitary gland

The pituitary gland is not a part of the brain. It is located below the hypothalamus, to which it is attached. Its function is to produce numerous hormones.

Nervous system disorder

Parkinson's disease

Cause

Parkinson's disease is a disorder of the nervous system. It is caused by the failure (for reasons unknown) to produce a neurotransmitter called **dopamine** in a part of the brain.

Symptoms

The failure to produce dopamine results in the inability to control muscle contraction. This results in symptoms such as:

- Trembling of the hands and/or legs
- Stiff and rigid muscles and body
- A shuffling unbalanced gait when walking
- A fixed, unblinking stare
- Difficulty with everyday activities such as dressing, washing and eating.

Thought processes are not affected until late in the course of the disease. Parkinson's disease is normally found in the elderly and is more common in men than in women.

Prevention

There is no known way to prevent Parkinson's disease.

Treatment

Initial treatment involves physiotherapy and special exercises along with the provision of special aids and home help to patients. Treatment with drugs to stimulate or mimic dopamine can reduce the symptoms but cannot stop the degeneration of the brain. Recent research suggests that electrical stimulation or implanting dopamine-secreting tissue (stem cells) into the brain may prove beneficial.



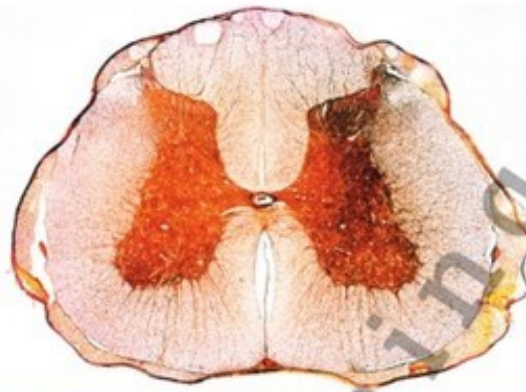
10.15 X-ray of the brain: electrodes are visible as a treatment for Parkinson's disease

Spinal cord

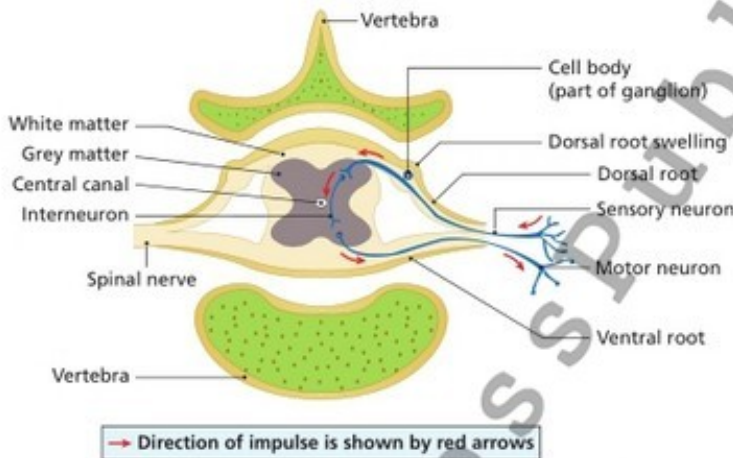
The spinal cord is composed of nerve tissue. It is surrounded by bony vertebrae, which protect it. The spinal cord transmits impulses to and from the brain. It also controls many reflex actions.

The spinal cord is located in the neural canal of the vertebrae. The neural canal is lined by the meninges.

In cross-section the spinal cord appears as an outer ring of **white matter** (axons only) surrounding an inner H-shaped region of **grey matter** (cell bodies). At the centre of the grey matter is the central canal, which contains cerebrospinal fluid.



10.16 Section through the spinal cord. the grey matter (red) is surrounded by white matter



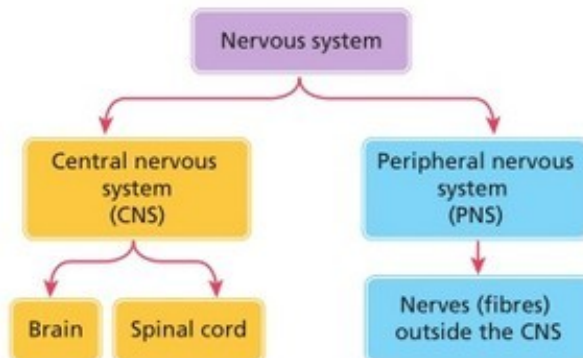
10.17 Transverse section (TS) of the spine

The dorsal root carries sensory neurons into the spinal cord and the ventral root carries motor neurons out. These neurons are usually linked by numerous interneurons in the grey matter. The dorsal root swellings contain ganglia, i.e. groups of cell bodies of sensory neurons. The dorsal and ventral roots combine to form 31 pairs of spinal nerves, which take impulses to and from the spinal cord.

Peripheral nervous system

The peripheral nervous system (PNS) mostly consists of nerve fibres outside the brain and spinal cord. These are made up of long dendrites or axons taking impulses to or from the CNS.

- The cell bodies of sensory nerves are located in ganglia in the PNS (i.e. in the dorsal root ganglia just outside the spinal cord).
- The cell bodies of motor neurons are found in the CNS (i.e. in the grey matter of the brain and spinal cord).



10.18 Parts of the nervous system

A reflex action is an automatic, involuntary, unthinking response to a stimulus.

Reflex action

The simplest form of activity in the nervous system is a reflex action.

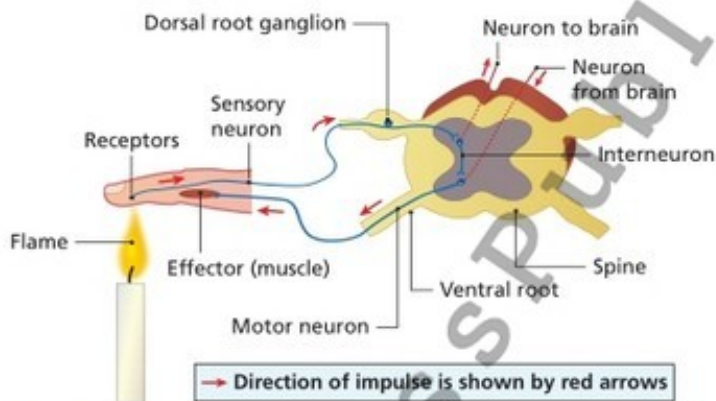
Examples of reflex actions

Many of the activities of the body are reflex and are controlled by reflex arcs. Examples include:

- The grasp reflex in children
- The movement of the iris of the eye
- Blinking the eyes for protection
- Breathing
- Control of blood pressure
- The protective actions we take when falling
- The knee jerk.



10.19 The knee jerk: an example of a reflex action



10.20 A reflex arc

Advantages of reflex actions

The advantage of reflex actions is that they are fast responses and so can **protect** the body from damage.

This is best understood by considering a relatively simple, 3-neuron withdrawal reflex, such as pulling the hand back from a hot flame.

Reflex arc

A reflex arc is the unit of function of the nervous system.

Withdrawal reflex

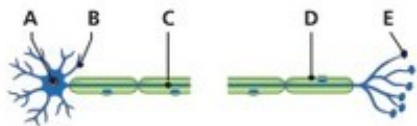
- 1 Receptors in the finger are stimulated by the hot flame.
- 2 Sensory neurons carry an impulse into the spinal cord through the dorsal root.
- 3 In the spinal cord numerous synapses are made with other neurons.
 - (i) An interneuron carries the impulse across the spinal cord to a motor neuron.
 - (ii) Another neuron takes the impulse up to the brain.
- 4 Motor neurons take the impulse straight out of the spine (through a ventral root) to the effector (i.e. a muscle or gland). This causes us to pull our hand back from the hot flame.
- 5 At the same time as the hand withdraws, the impulse reaches the brain. This makes us aware of what has happened and we feel some pain. However, the action was involuntary, it was not controlled by the brain.

This account is a simplification of the process. Extra connections and pathways exist to add further complexity to the process. For example, we pull our arm **up** from a hot cooker, but **down** when changing a hot overhead bulb. Also, reflex actions can be inhibited, e.g. we drop a hot plate but we attempt to place an expensive hot dish down gently.

A reflex arc is the pathway taken by a nerve impulse in a reflex action.

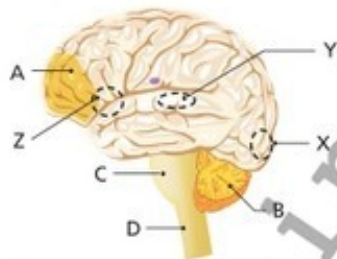
Questions on Module 10

- 1 (a) Name the three types of neurons.
 (b) Distinguish between the three types of neurons in terms of where they carry impulses to and from.



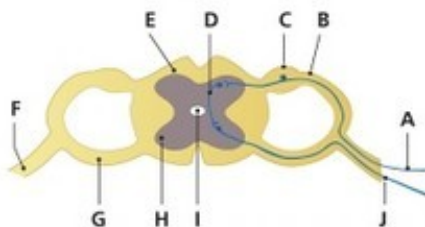
10.21 Neuron

- 2 (a) Name the type of neuron shown in diagram 10.21.
 (b) Name, and give one function for, all the parts labelled A to E.
 (c) In what form does the impulse travel along structure C?
 (d) In what direction does the impulse travel at C?
 (e) Name the other two types of neurons and draw similar labelled diagrams for each.
- 3 (a) What is a synapse?
 (b) Give two benefits of synapses.
- 4 Describe how an impulse arriving at one side of a synapse can continue across the synapse.
- 5 (a) What is the function of neurotransmitters?
 (b) Name two neurotransmitters.
- 6 Distinguish between:
 (a) The central and peripheral nervous systems
 (b) Afferent and efferent neurons
 (c) Dendrites and axons
 (d) Synapse and synaptic cleft.
- 7 (a) Name the parts of the CNS labelled A, B, C and D in diagram 10.22.
 (b) Give two functions for each part named in answer (a).
 (c) Assign each of the following abilities to one of the regions labelled X, Y or Z: eyesight, speech, hearing.



10.22 Central nervous system (CNS)

- 8 Give a location and function for each of the following:
 (a) Meninges
 (b) Cerebrospinal fluid
 (c) Corpus callosum
 (d) Hypothalamus
 (e) Pituitary gland.
- 9 (a) Name a disorder of the nervous system.
 (b) Give three symptoms of this disorder.
 (c) Name one way in which the disorder may be treated.
- 10 (a) Name the parts labelled A to J in the TS of the spinal cord shown in diagram 10.23.



10.23 Cross-section of spinal cord

- (b) Describe the direction of the impulse in each of the structures labelled A, D and J.
 (c) Name the substance in the part labelled I.
- 11 (a) Give three examples of reflex actions.
 (b) What is the advantage of a reflex action?

- 12 Complete the gaps to show the neurones involved in a nervous response.
- receptor •
 - central nervous system
 - • muscle
- 13 The pufferfish is a type of fish which produces a toxin inside some organs of its body. Some people eat pufferfish, which must be carefully prepared to avoid the toxin being in the food. The toxin works by blocking action potentials in neurones. Eating the toxin is fatal. Suggest two symptoms which could be caused by eating small quantities of this toxin.
- 14 "A stronger stimulus to a sensory receptor will result in a larger action potential." Explain whether or not this statement is correct.
- 15 Vertebrates have myelinated sensory and motor neurones, but invertebrates have neurones without any myelin sheath. Suggest why vertebrates need myelinated neurones but invertebrates do not.
- 16 Explain why a severe injury to the lower back may result in loss of movement of the legs, but have no effect on the upper body or arms.
- 17 Explain why the reaction time of a reflex response is faster than that of a voluntary response.

Module 11 The skeleton and muscles

Learning objectives

- Describe the structure and function of the human skeleton
- Describe the classification, function and location of different types of joints
- Explain the role of ligaments, tendons and cartilage in the human musculoskeletal system
- Investigate the ultrastructure of striated muscle [10.1.6.1]
- Explain the mechanism of muscle contraction [10.1.6.2]
- Establish the relationship between fast and slow muscle fibres [10.1.6.3]
- Investigate the application of biomechanics in robotics [10.4.4.1]
- Explain the mechanism of cardiac automatics using an electrocardiogram [10.4.4.2]

Introduction

The skeletal and muscular systems work together in most animals to form the musculoskeletal system. This system is controlled by the nervous system.

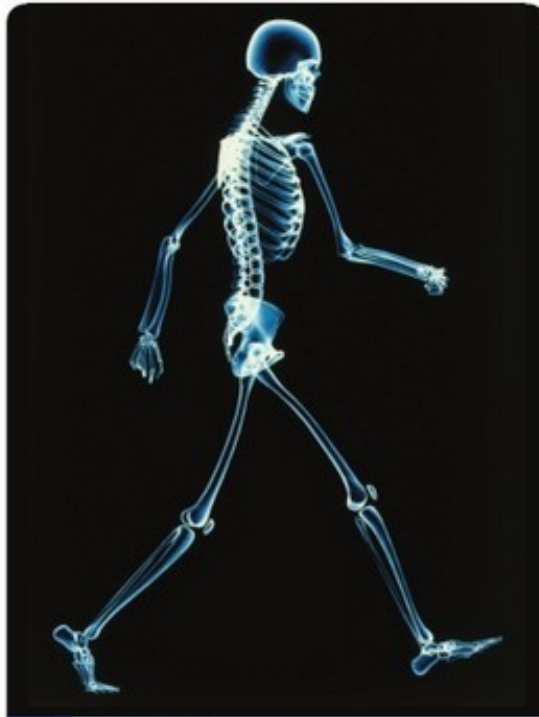
Muscles associated with the skeleton are called skeletal muscle. Other types of muscles are found in the heart (cardiac muscle) and in places such as blood vessels, intestines, the bladder and the uterus (smooth muscle).

Functions of the skeleton

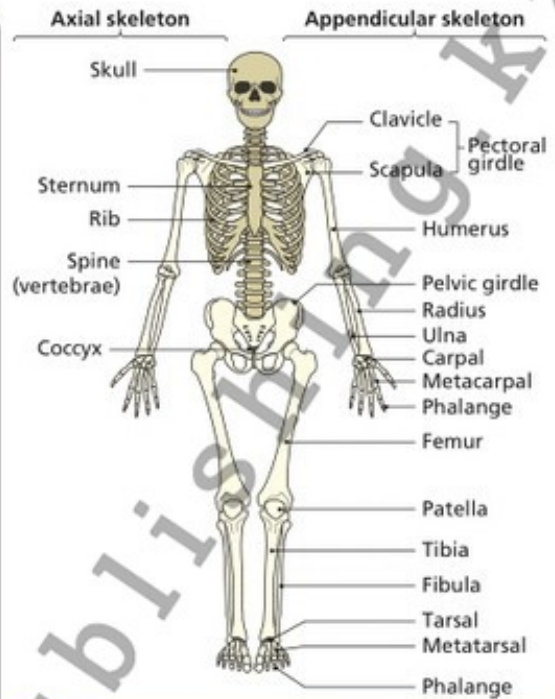
- **Support:** the bones of the skeleton provide a rigid framework that holds the body upright.
- **Protection:** the skull protects the brain, the vertebrae protect the nerves of the spinal cord and the ribs protect the heart and lungs.
- **Movement:** bones provide a system of rigid levers against which muscles can pull. Without rigid bones, movement would not be possible.
- **Shape:** the shape of the body is determined to a large extent by the skeleton. If a person has long bones they are tall; the bones in the feet determine the width of the foot.
- **Manufacture of blood components:** bone marrow makes red blood cells, white blood cells and platelets.

Structure of the human skeleton

The adult human skeleton has 206 bones and is divided into the axial and appendicular skeletons.



11.1 The human skeleton



11.2 The human skeleton, showing the parts of the axial skeleton and the appendicular skeleton



The axial skeleton consists of the skull, spine, ribs and sternum (breastbone).

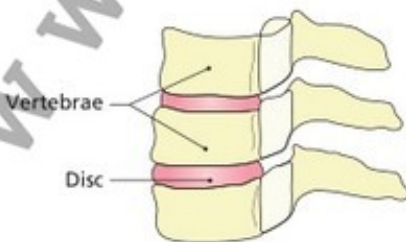
Parts of the axial skeleton

Skull

The skull, or cranium, consists of over 20 bones fused together.

Spine

The spine is made of 33 bones called **vertebrae**. These are arranged into five regions, as shown in diagram 11.3. The top 24 vertebrae are held together by ligaments and can **move slightly** relative to one another. They are separated by (intervertebral) discs of cartilage. These discs have a hard outer layer and a soft, jelly-like centre. They act as shock absorbers and protect the vertebrae.



11.4 The position of the discs in relation to the vertebrae

Region	Number of vertebrae
Cervical (neck)	7
Thoracic (chest)	12
Lumbar (back)	5
Sacrum (hip)	5
Coccyx (tail)	4



11.3 Regions of the spine and the number of vertebrae

The last nine vertebrae are **fused** together and there are no discs between them. No movement occurs between these vertebrae. Together they form the sacrum and coccyx.



Sometimes the soft centre of a disc bulges out and compresses some spinal nerves. This may result in pain in the back or leg (i.e. the region to which the nerve is attached). This condition is often called a 'slipped disc'.

In addition it is known that people are taller in the mornings when their discs are fully expanded. During the day the discs become compressed (due to gravity) and people become slightly shorter.

Vertebrae have different shapes, depending on where they are located in the spine. The general shape of a vertebra is shown in diagram 11.5.

Rib cage

The rib cage consists of the sternum (breastbone) and 12 pairs of ribs. All ribs are attached to the vertebrae of the spine.

- The top seven ribs are attached to the breastbone at the front of the body. They are called **true ribs**.
- The next three ribs (i.e. numbers 8, 9 and 10 from the top) are attached to each other at the front of the chest by cartilage. They are called **false ribs**.
- The bottom two ribs, called **floating ribs**, are only attached to the spine (i.e. they do not attach to anything at the front of the body).

Parts of the appendicular skeleton

Pectoral girdle

The pectoral girdle forms a connection with the vertebral column and with the arms (i.e. the humerus, radius, ulna, carpals, metacarpals and digits (fingers), which contain the phalanges).



The **appendicular skeleton** is composed of the limbs (arms and legs), the pectoral (shoulder) girdle and the pelvic (hip) girdle. The **pectoral girdle** consists of the collarbone (or clavicle) and the shoulder blade (or scapula).

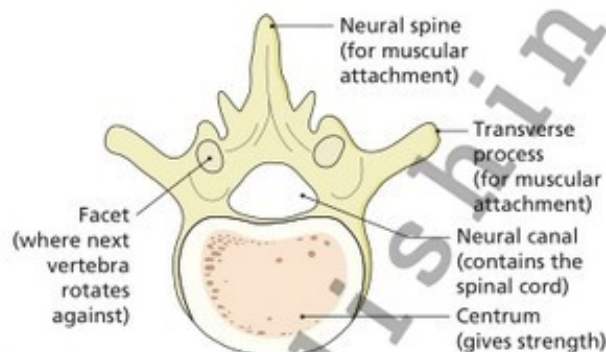
Pelvic girdle

Each half of the pelvic girdle consists of three fused bones. The two halves are joined by a band of flexible cartilage.

The pelvic girdle is fused (joined firmly) to the spine (at the sacrum). The hollow cavity where the hip bones attach to the sacrum is called the pelvis. The pelvic girdle is also connected to the legs (the femur, patella, tibia, fibula, tarsals, metatarsals and digits (toes), which contain the phalanges).



The **pelvic girdle** is composed of two halves of the hip joined to the sacrum.



11.5 Transverse section (TS) of a typical vertebra



11.6 Rib cage (CT scan): note that cartilage (purple) is found at the ends of many ribs

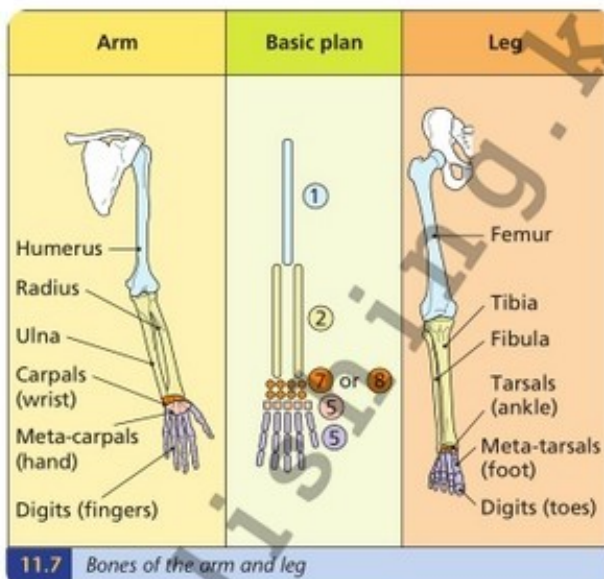
Limbs

The arms and legs have a similar design pattern, as shown in diagram 11.7.

Each limb ends in five digits (fingers or toes). For this reason they are called **pentadactyl limbs**. The phalanges are the individual bones of the fingers and toes. Each finger and toe has three phalanges, except the thumb and big toe, which only have two phalanges.



An important feature of the limbs of great apes (orangutan, gorilla and chimpanzee) and humans is that they have opposable thumbs. This means that the thumb can be pushed against all the other four digits. This gives much greater powers of grip and manipulation.



11.7 Bones of the arm and leg

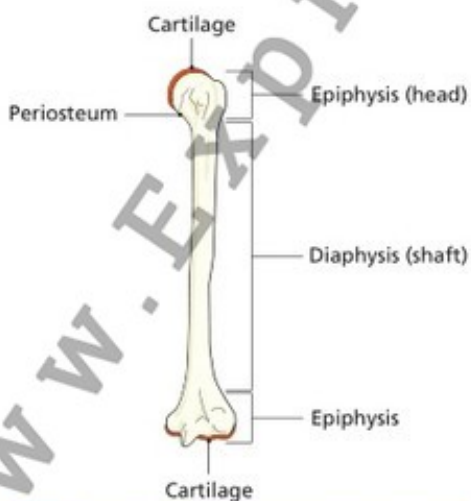
Cartilage

Cartilage contains a firm but flexible fibrous protein called collagen. Cartilage is lacking in blood vessels and nerves. For transport it depends on materials diffusing through to the cells that form it (compared with bone, which has a rich blood supply). This is why cartilage is slower to heal than bone.

Cartilage is found in the pinna of the ear, the nose, the trachea and in the discs between the vertebrae. Cartilage also covers the ends of bones.

Function of cartilage

Cartilage protects bones (by acting as a shock absorber) and allows friction-free movement.



11.8 External structure of a long bone

Structure of long bone

External structure

Long bones such as the femur are enclosed by a membrane called the periosteum. This membrane contains blood vessels and nerves. The long shaft of a bone is the diaphysis and the head of a bone is called the epiphysis.

Internal structure

The inside of a bone has three main regions: compact bone, spongy bone and the medullary cavity.

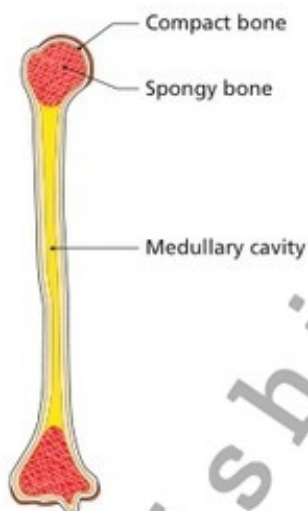
Compact bone

Compact bone is made of bone-forming cells embedded in a surrounding medium or matrix. The matrix is composed of 70% inorganic (non-living) salts such as calcium phosphate and 30% protein (called collagen). Bone cells are supplied with nutrients by blood vessels. Nerve fibres also run throughout a bone.

Compact bone is mostly found in the shaft (diaphysis) of a bone. It is also located as a layer around the end (epiphysis) of a bone.

Function of compact bone

The calcium salts give compact bone its strength and protein gives bone its flexibility. Bone cells and protein are both organic (living) materials.



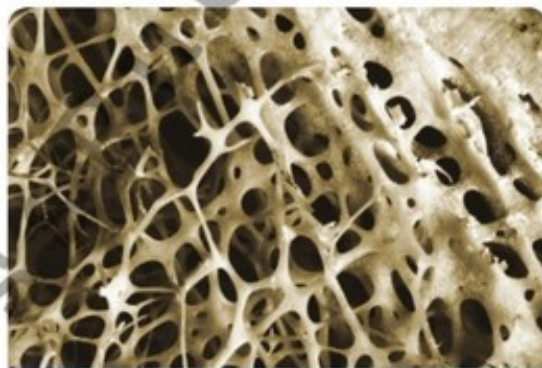
11.9 Internal structure of a long bone

Spongy bone

Spongy bone is like compact bone that contains numerous hollows. Spongy bone consists of a network of thin, bony bars separated by different-sized spaces. The spaces in spongy bone are filled with red bone marrow, which produces blood cells. Spongy bone is found mostly in the ends (epiphyses) of bones.

Function of spongy bone

Spongy bone makes blood cells and also gives strength and rigidity to the skeleton.



11.10 Spongy bone: note that the red marrow is not visible in this scanning electron micrograph (SEM)

Medullary cavity

The medullary cavity contains bone marrow.

- In young people, bone marrow is full of active, red marrow. Red marrow makes blood components.
- In adults, active marrow is confined to the spongy bone. The medullary cavity of adults contains inactive, yellow, fat-rich marrow. This marrow can convert to red marrow if the body requires increased blood cell formation.

Renewal of bone

Continual renewal of bone is dependent on physical activity, hormones and diet.

- When bones are stressed by physical activity they become thicker and stronger (the osteoblasts are stimulated). This happens especially at the sites where muscles attach to the bone. Lack of stress on bones causes them to become thin.
- The main hormones affecting bone development are growth hormone, sex hormones and parathormone.

- ▶ Growth hormone and many sex hormones increase the size of bones. This can be seen clearly at puberty when bone mass in the body may increase rapidly.
- ▶ Parathormone removes calcium from bone. This happens so that the level of calcium in the blood can be raised (a constant level of calcium in the blood is essential for muscles and nerves to work properly). It is essential to have sufficient supplies of calcium in the diet to replace the calcium lost from bones.



Osteoporosis is the loss of protein (collagen) material from bone. It causes bones to become brittle and easily broken. It should not be confused with osteomalacia, which is the loss of minerals (calcium) from bone due to the lack of vitamin D.



11.11 A normal bone (left) and a bone showing osteoporosis

Joints

Joints may be classified according to the degree of movement they allow.



A **joint** is where two or more bones meet.

Immovable joints

Immovable (fixed or fused) joints include the skull and pelvic girdle. The junction between fused bones is called a suture. These joints provide strength, support and protection.

Slightly movable joints

The joints between the vertebrae in the upper spinal column are slightly movable. In these joints the bones are separated by a disc of cartilage and the bones are held in place by ligaments. These ligaments limit the amount of movement possible in order to protect the nerves of the spinal cord.

Freely movable (synovial) joints

In synovial joints, the ends of the bones are covered with cartilage and the bones are separated by a cavity. The bones are held in place by ligaments, which prevent excessive movement of bones at joints.

These joints are enclosed in a synovial membrane. The membrane secretes synovial fluid, a clear sticky liquid resembling egg white. This fluid lubricates the joint and reduces friction in the joint.

Examples of synovial joints include ball-and-socket joints and hinge joints.



Synovial fluid is produced in movable joints to lubricate and reduce friction.



11.12 An x-ray of a hip joint

- **Ball-and-socket joints** (e.g. the shoulder and hip) allow movement in all directions. They are unable to support heavy loads.
- **Hinge joints** (e.g. the elbow and knee) allow movement in one direction only. They can support heavy loads.



Excessive fluid in joints often occurs because of an injury. As a result of the injury, the synovial membrane secretes more fluid and the joint swells. The usual treatment for excess fluid is summarised by the word RICE (i.e. rest; ice or cold treatment applied as soon as possible; compression with a bandage or sock; elevation of the limb to allow the fluid to drain away). Occasionally anti-inflammatory drugs may be needed.



Ligaments are strong, fibrous, slightly elastic tissues that connect bone to bone.

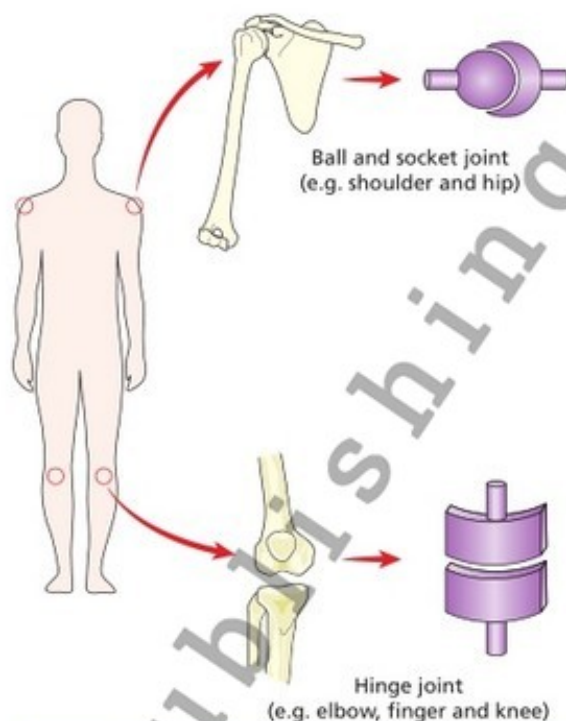
Tendons are strong, flexible, inelastic fibres that connect muscle to bone.

Ligaments

Ligaments are more flexible when warm; hence the need for warming-up exercises before physical activities, to prevent ligaments from being damaged.

Tendons

Tendons are mostly composed of collagen and contain some blood vessels.



11.13 Types of synovial joint



11.14 A typical synovial joint (the knee)

Musculoskeletal disorders

Arthritis

Cause

Arthritis is a skeletal disorder resulting from inflammation (swelling) of a joint. There are over 100 types of arthritis.

The most common type of arthritis is **osteoarthritis**. This usually occurs from 50 years of age onwards. It is caused by the cartilage in synovial joints wearing down. The underlying bones enlarge and more synovial fluid forms. The joint(s) become sore and stiff.

Rheumatoid arthritis is the most severe form of joint inflammation. It is caused genetically by the body's immune system turning on itself (i.e. it is an auto-immune disease).

The synovial membranes are attacked first. The joint swells and in time may become damaged and deformed. It can occur at any age.

Prevention

As osteoarthritis is caused by wear and tear on the cartilage in joints, it may be prevented by reducing damage to joints. This may involve using proper footwear when running, avoiding running on hard surfaces (especially roads) and perhaps exercising by walking or swimming instead of running.

Treatment

There is no cure for either form of arthritis. Treatments include rest, exercises to maintain mobility and strength, weight loss, anti-inflammatory medications, steroids, drugs to reduce the immune response and possibly surgery to replace the joint.



11.15 The effects of rheumatoid arthritis

Muscles

There are three types of muscle: skeletal, smooth and cardiac muscle.

Skeletal muscle

Skeletal muscle is also called striated, striped or voluntary muscle. There are over 600 skeletal muscles in the body, and they make up 50% of body weight.

Skeletal muscle is concerned with body movements. It can contract quickly, but tires very easily (try opening and closing your fist for 1 minute!). This type of muscle is under **voluntary** or conscious control.

Smooth muscle

Smooth muscle is also called unstriped or involuntary muscle. It is found in internal structures such as the digestive system, blood vessels, bladder and uterus. It contracts slowly and is slow to tire. It is under **involuntary** or unconscious control.

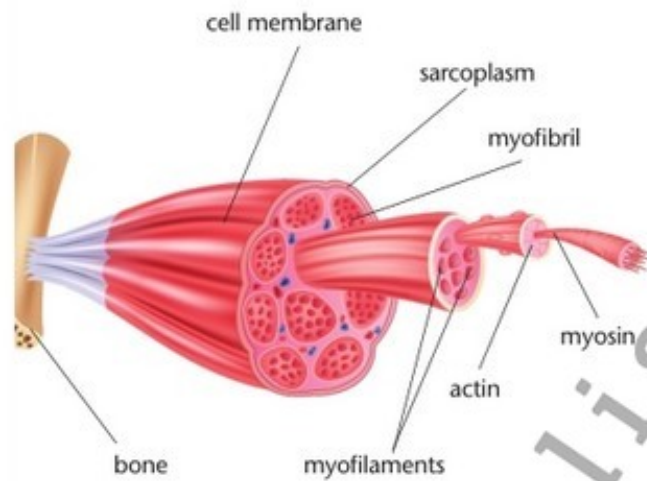
Cardiac muscle

Cardiac muscle is found in the heart. It has many mitochondria, contracts strongly and **does not tire** as easily as skeletal muscle. It is **involuntary**, i.e. not under conscious control.

Structure of striated muscle

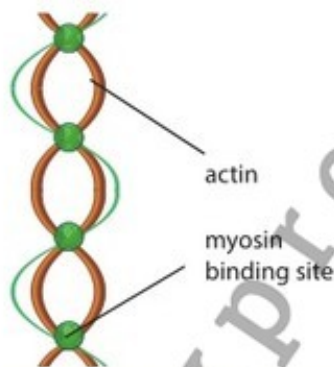
The cells in muscle fibres contain many nuclei, mitochondria - the source of ATP for contraction - and sarcoplasmic reticulum which contains calcium ions. The cell membrane of a muscle fibre is called the sarcolemma. The sarcolemma has folds which fold inwards across the fibres to allow electrical impulses to spread out through the sarcoplasm.

Each muscle is called a fibre. Each fibre consists of a bundle of myofibrils. Myofibril is made of two myofilaments - actin and myosin.



11.16 A muscle fibre

Actin is thinner than myosin and appears under a microscope as a light band. Myosin, appears as a dark band. Actin consists of 2 threads wrapped around each other. At each twist there is a binding site for myosin. In a relaxed state, a molecule called tropomyosin covers these sites.



11.17 Actin



11.18 Myosin

The myosin filament consists of many myosin molecules. Each molecule has a tail and a double globular head. The head attaches to the myosin binding sites on the actin where the actin and myosin filaments overlap. The heads contain ATPase enzyme which releases energy from ATP to power muscle contraction.

Contraction occurs when an impulse from a motor neurone reaches the synapse at the junction with the muscle. ATP attached to the myosin heads cause them to flex and attach to the actin in the overlapping areas. ATP is hydrolysed to ADP + P and the energy released causes the heads to alter their angle to their tails and this pulls the actin filament past the myosin filament. When there is no further stimulation, the tropomyosin moves back to cover the actin binding sites.

Antagonistic pairs

Muscular contraction is an active process and requires energy in the form of ATP. Muscles are connected to bones by tendons. When the muscle contracts, the tendon pulls on the bone, causing it to move.

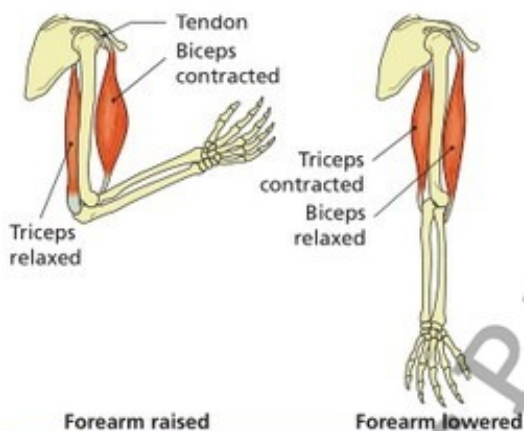
Muscles can only pull (by contracting or shortening).

They cannot push. For example, in the forearm a muscle on top of the humerus called the **biceps** contracts to pull the lower arm up.

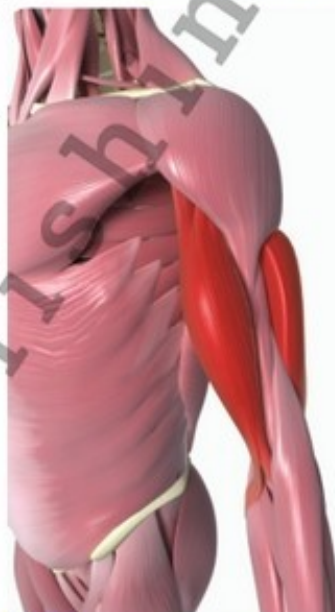
A second muscle is required to straighten the lower arm.

This is a smaller, weaker muscle (called the **triceps**), located at the back of the humerus. It contracts to straighten the arm.

An antagonistic pair is two muscles that have opposite effects to each other.



11.19 An antagonistic pair in the arm



11.20 A model showing the biceps (larger) and triceps

In this example, the biceps is a flexor (i.e. it closes the joint) and the triceps is an extensor (it opens the joint).



Generally flexors are stronger than extensors. The palm of the hand has many flexors (i.e. the front of the hand is fleshy), but the back of the hand has smaller, weaker extensors (the back of the hand has less 'padding'). The same effect is seen on the back and front of the shins and thighs.

Fast and slow twitch muscle fibres

Muscles consist of two different types of fibre: slow twitch muscle fibres and fast twitch muscle fibres. Slow twitch fibres contract slowly but work for extended periods without tiring. Fast twitch muscles contract quickly but use up a lot of energy in the process.

The majority of muscles consist of both types of fibre but certain muscles only contain one type of muscle fibre. The muscles responsible for eye movement are made up of fast twitch muscle fibres whereas the soleus muscle in the lower leg contain slow twitch fibres.

The primary difference between these fibres is that slow twitch fibres are red because the energy they need to contract uses oxygenated blood. Fast twitch muscles do not use oxygen for the energy used in contraction and have a lighter colour. Slow twitch muscles fibres typically support endurance activities such as jogging or cycling whereas fast twitch muscles fibres are deployed when greater speed and force of movement is required and, therefore, tire more quickly and need longer phases of rest after exertion.

Cardiac muscle

Control of heartbeat

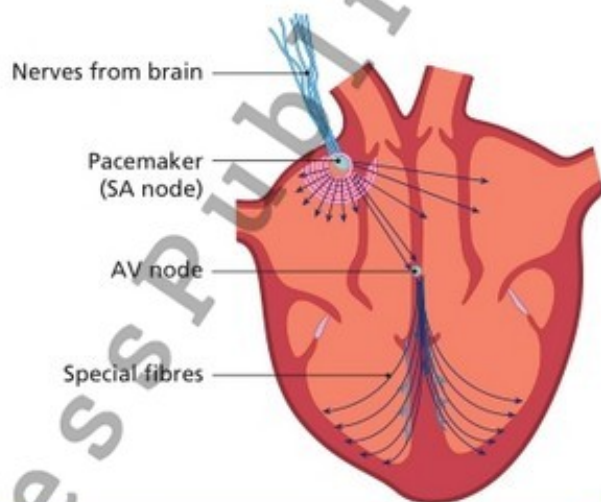
If a heart is removed from a body and kept in a nutritive, oxygen-rich fluid it will continue to beat. This shows that heartbeat can occur independently of the brain.

Heartbeat is controlled by a small bundle of specialised tissue called the **pacemaker**. This is located in the wall at the top of the **right atrium**. The pacemaker sends out regular electrical impulses, which initially cause the atria to contract. These impulses then cause the ventricles to contract. This means that blood is first pumped from the atria to the ventricles and a split second later it is pumped from the ventricles out of the heart.

The frequency of these impulses can be increased or decreased by the brain (i.e. the brain can cause the pacemaker to speed up or slow down the rate of heartbeat).

Heartbeat is controlled in the following way:

- 1 The pacemaker (also called the SA, or sinoatrial node) pulses and causes the atria to contract.
- 2 The electrical impulse from the pacemaker stimulates the AV (atrioventricular) node. This is similar to the pacemaker but is located further down in the right atrium near to where it joins the ventricle.
- 3 The AV node sends an impulse down special muscle fibres located in the septum.
- 4 The impulse is passed out to the walls of the ventricles by thin fibres. The impulses from these fibres cause the ventricles to contract.



11.21 Path of nerve impulses in the heart

The pacemaker controls the rate of heartbeat. However, nerves from the brain (along with hormones) can change the rate at which the pacemaker (and therefore the heart) operates.

- Factors that increase the rate of heartbeat include exercise, temperature, emotions and shock.
- Factors such as relaxation, sleep and alcohol decrease the rate of heartbeat.

Diastole is when the heart chambers relax.
Systole is when the heart chambers contract.

The stages of heartbeat (cardiac cycle)

The events that take place during one heartbeat occur in three stages.

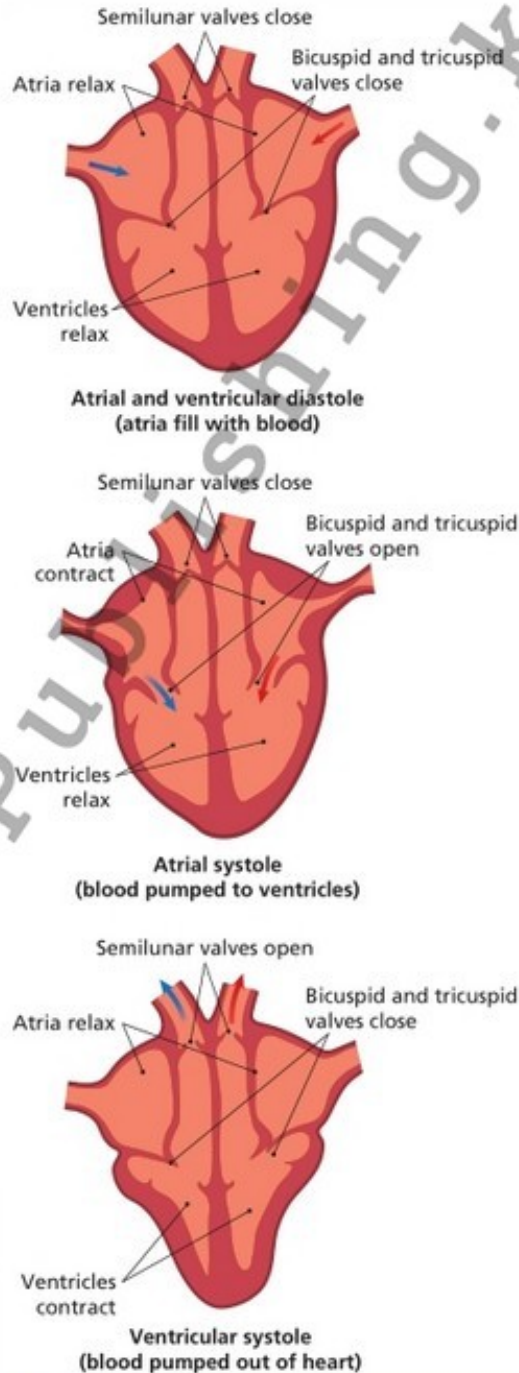
- 1 **Blood enters the heart:**
 - The atria and ventricles are both relaxed (diastole)
 - Blood enters the atria
 - All valves are closed.

- 2 Blood is pumped from the atria to the ventricles:
 - Electrical impulses from the pacemaker cause the atria to contract (atrial systole)
 - Blood is pumped to the ventricles
 - The tricuspid and bicuspid valves open
 - The vena cava and pulmonary veins close to stop blood entering the atria
 - The semilunar valves remain closed.

- 3 Blood leaves the heart:
 - The atria relax
 - Impulses from the AV node cause the ventricles to contract (ventricular systole)
 - Blood is forced out of the heart into the pulmonary artery and the aorta
 - The pressure forces the semilunar valves to open
 - The pressure closes the tricuspid and bicuspid valves
 - The ventricles now relax again
 - The semilunar valves close, which prevents blood from flowing back into the heart (or ventricles)
 - The vena cava and pulmonary veins open and the cycle starts again.



The characteristic double sound of heartbeat (called the 'lub-dub' sound) is caused by the valves being forced shut. The low-pitched, quieter, longer-lasting 'lub' sound is due to the bicuspid and tricuspid valves being forced shut when the ventricles contract. The higher pitched, louder, much shorter 'dub' sound is due to the semi-lunar valves snapping shut. A heart murmur is any abnormal sound associated with heartbeat. A heart murmur may indicate damage to one or more of the valves.



11.22 Blood flow through the heart

Electrocardiograms



11.23 An electrocardiogram (ECG) test

An electrocardiogram (ECG) test is a relatively straightforward procedure to check the rhythm and electrical activity of the heart. It is used to detect the electrical signals produced by the heart each time it beats.

The conditions an ECG is typically used to help diagnose are:

- **arrhythmias** – a heart beating too slowly, too quickly, or irregularly
- **coronary heart disease** – the blood supply to the heart being blocked or interrupted by a build-up of fatty substances
- **heart attacks** – the supply of blood to the heart being suddenly blocked
- **cardiomyopathy** – the thickening or enlargement of the heart walls

In a healthy adult heart at rest the SA node in Figure 11.23 sends an electrical signal to begin a new heartbeat 60 to 100 times a minute. The signal moves from the right and left atria causing the atria to contract and moving blood into the ventricles the lower chambers of the heart. On an ECG the electrical signal moving through the atria is recorded as the P wave (Figure 11.23).

The electrical signal then passes between the atria and ventricles through a group of cells called the atrioventricular (AV) node. It slows down as it passes through the AV node, allowing the ventricles time to finish filling with blood. This stage of the process is the flat line between the end of the P wave and the beginning of the Q wave.

The electrical signal then leaves the AV node and travels through a collection of heart muscle cells used in electrical conduction called His. It then travels into the right and left bundle branches, spreading quickly across the heart's ventricles. This causes them to contract and pump blood to the lungs and around the rest of the body. On the ECG this is seen as the QRS waves.

The ventricles then recover their normal electrical state (recorded as the T wave on the ECG). The muscle stops contracting to allow the heart to refill with blood. This whole process is repeated for each new heartbeat.

Advances in technology and ways of recording data mean that ECGs can be done in a range of ways. An ambulatory ECG, for example, involves attaching a small device to the waist to monitor your heart condition over a number of days as you move around.

Biomechanics

Biomechanics is the study of the mechanics of a living body. It explores the forces exerted by muscles and gravity on the skeletal structure of animals. The focus of study is usually on the mechanics of a part of body or the function part of the musculoskeletal system e.g. limbs, joints and muscle fibres and how they impact movement in areas such as sport or heart muscles in medicine.

Robotics

Robotics is an area of study of artificial intelligence that focuses on the development of systems and devices that can move and respond to sensory input.

Biological Robotics

Biological robotics is a field of research that uses robotics:

- 1 to help understand and model biological sensorimotor systems or
- 2 to create innovative robotic devices that can help humans

Robots like humans:

- rely on a large variety of sensors
- require a neural control system
- use actuators or muscles
- are limited by their mechanical structure

Biological robotics looks at these components together, considering these different features as part of an action-perception loop.

Applications of biological robotics

- development of sensors based on biological counterparts
- development of versatile robots with novel capabilities
- development of innovative robotic tools
- development of robots for working in hazardous environments or performing routine tasks
- development of robotic systems to support the physically or neurologically impaired

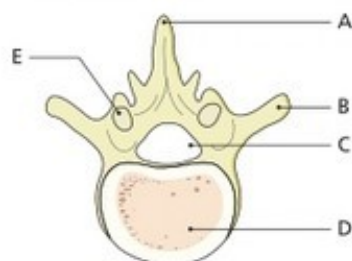


11.24 A robotic prosthetic arm

Research a product or system related to one of the above areas of application to present to the class. Your presentation must show the innovation is related to or inspired by a biomechanical or neurological system.

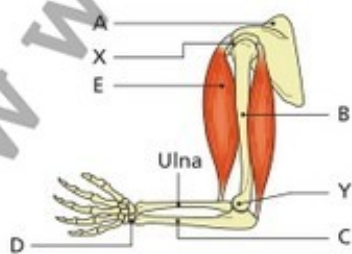
Questions on Module 11

- State the functions of the skeleton.
 - Explain briefly how each function is carried out.
- Name the parts of:
 - The axial skeleton
 - The appendicular skeleton.
- Name the parts labelled A to D in the TS of a vertebra shown in diagram 11.25.



11.25 Transverse section of a vertebra

- What is the common function of the structures labelled A and B?
 - What is found in C during life?
 - What is the function of E?
- Distinguish between:
 - True, false and floating ribs
 - Pectoral and pelvic girdles
 - Compact and spongy bone
 - Ligaments and tendons
 - Red and yellow marrow.
 - What are vertebrae?
 - Name the regions of the spine and say how many vertebrae are located in each region.
 - Name the structures located between the vertebrae.
 - What is the function of these structures?
 - Name the bones labelled A, B, C and D shown in diagram 11.26.



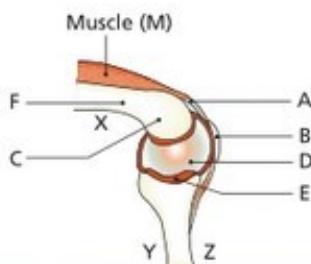
11.26 Joints and muscles in the arm

- What types of joint are found at X and Y?
 - Name the muscle labelled E. Say if this muscle is contracted or relaxed.
- How many ribs are normally present in a complete rib cage?
 - Outline the attachment of the ribs:
 - At the back
 - In the chest region.
 - Diagram 11.27 represents the hip.



11.27 Hip joint

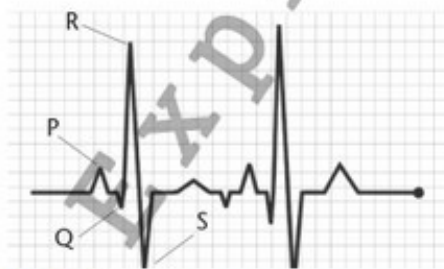
- Name the parts labelled A to D.
 - What function is common to parts B and C?
 - What type of joint is represented by the diagram?
- Draw a long section of a bone to show the positions of:
 - Compact bone
 - Spongy bone
 - Medullary cavity
 - Red marrow
 - Periosteum
 - Epiphysis
 - Diaphysis.
 - Name the parts labelled A to F in diagram 11.28 of the knee.



11.28 Knee joint

- What type of joint does the knee represent?

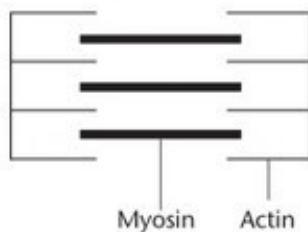
- (c) Is the muscle, labelled M, a flexor or extensor? Explain your answer.
- (d) Is the antagonistic partner of muscle M located at position X, Y or Z?
- 11 Give one function for each of the following:
- (a) Cartilage (b) Ligament
(c) Tendon (d) Synovial fluid.
- 12 (a) What is arthritis?
(b) Name and distinguish between two types of arthritis.
(c) Name two methods by which the effects of arthritis may be relieved.
- 13 With regard to heartbeat, answer the following:
- (a) What structure causes it?
(b) Where is this structure located?
(c) What effect has exercise on heartbeat?
(d) How is heartbeat normally measured?
- 14 (a) Distinguish between systole and diastole.
(b) What is meant by blood pressure?
(c) Suggest one reason why high blood pressure is unhealthy.
- 15 (a) Explain what is happening in terms of the heartbeat at points P, Q, R, S and T in the ECG printout.



11.29 ECG printout

- (b) Name two heart conditions an ECG can help to detect.
- 16 How is biological robotics different to biomechanics?
- 17 Describe the similarities and differences between the parts of the skeleton that are found in the skull and those which are found in the arm.

- 18 State two pieces of evidence that show bone is living tissue.
- 19 Explain why obesity increases the risk of developing osteoarthritis in the hip and knee joints.
- 20 Describe how the movement at a hinge joint differs from that at a ball and socket joint and give one example of each in the human body.
- 21 (a) Describe the meaning of the term pentadactyl limb.
(b) Humans, horses, turtles, birds and frogs all have pentadactyl limbs. Explain why some biologists use this as evidence for evolution.
- 22 Explain, with reference to the skeleton, why animals like humans and monkeys can lift objects with their hands while animals like cats and dogs cannot do this.
You do **not** need to make reference to whether the animal walks on two or four legs.
- 23 When babies are born, many of their bones are formed completely from cartilage. The bones gradually harden as they get older, only being fully hardened at adulthood. Explain this change in bone composition with aging.
- 24 The diagram shows how myosin and actin microfilaments are arranged in an extended muscle.



11.30 Myosin and actin microfilaments

Complete the diagram below to show how myosin and actin are arranged when the muscle is contracted.



- 25 What are the roles of calcium ions and ATP in muscle contraction.

Module 12 Biotechnology

Learning objectives

- Discuss the advantages and disadvantages of using living organisms in biotechnology (10.4.3.1)
- Describe the importance of the use of DNA for taxonomy, medicine and solving crime (10.4.3.2)
- Explain the stages of genetic engineering manipulation (10.4.3.3)
- Discuss ethical issues in the use of GMO (10.4.3.4)

Introduction

Biotechnology is the use of living things, or parts of living things, to produce useful products.

The organisms used in biotechnology include plants, animals and microorganisms. In some cases, enzymes are used instead of entire organisms.

In particular, bacteria and fungi such as yeast are used to produce a vast range of useful products such as foods, drugs, alcohols, hormones and enzymes.

Old examples of biotechnology include the use of yeast to make alcohol or to raise the dough in bread-making. In the last century, fungi and bacteria were discovered and are now used to produce antibiotics.

In recent years microorganisms are being altered (by adding genes) to allow them to produce a huge range of new products.



12.1 Vats used to make drugs in biotechnology

Industrial uses of biotechnology

- Yeast is used in breweries to produce alcohol.
- Bacteria are used to produce special enzymes. These enzymes are added to washing powders and stain removers. Such products are often called biological detergents. The enzymes help to remove difficult stains caused by materials such as fat, inks and blood.

Medical uses of biotechnology

- Bacteria and fungi are used to make antibiotics.
- Altered organisms (such as bacteria and yeast) are used to produce medically important products such as:
 - ▶ hormones (e.g. insulin which is used to treat diabetes)
 - ▶ antibodies (which are used to treat infections)
 - ▶ chemicals needed to clot blood (these chemicals are used by haemophiliacs whose blood will not naturally clot).

Biotechnology allows these products to be produced cheaply, in huge quantities and without any dangerous side-products.

Antibiotics

Antibiotics are normally used to control bacterial infection, but they can treat some fungal diseases. Note that antibiotics do **not** affect viruses.

Originally antibiotics were isolated from fungi. Now antibiotics are mostly produced by genetically engineered bacteria.

Since the 1940s antibiotics have been widely used to treat bacterial infections. Many new antibiotics have been discovered, e.g. streptomycin, neomycin and tetracycline.



Pathogenic bacteria are bacteria that cause disease.

Antibiotics are chemicals produced by microorganisms that stop the growth of, or kill, other microorganisms without damaging human tissue.

Antibiotic resistance

When an antibiotic is used to treat a bacterial infection, most of the bacteria are killed. However, antibiotic-resistant bacteria have developed (and continue to do so) by mutations.

These bacteria are not affected by the antibiotic that is being used. This means that new antibiotics must be produced continually to treat newly resistant bacteria.

If a person is taking antibiotics, then all the bacteria in that person are killed. If one antibiotic-resistant bacterium arises by mutation or enters that person's body, then this resistant bacterium has no competitors.

The gene for antibiotic resistance is usually located on a plasmid. Bacteria can pass copies of their plasmid onto other bacteria. In this way antibiotic resistance can pass from one bacterium to another.

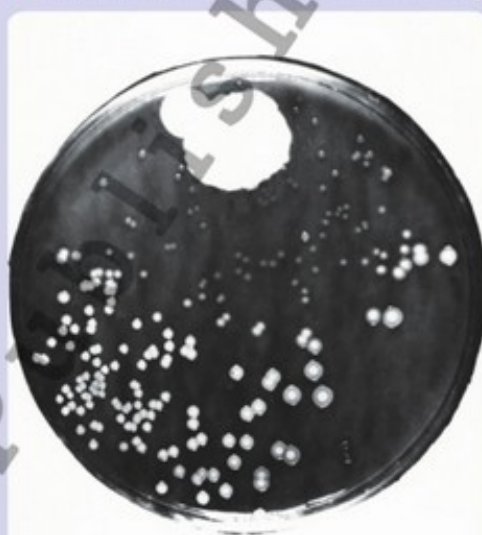
The antibiotic-resistant bacteria can reproduce very fast and take over a person's body. The person will then develop an infection for which the antibiotic is not an effective treatment.

In recent times bacterial strains have emerged that are resistant to almost all known antibiotics. These bacteria are said to be **multi-resistant**. Examples of such 'superbugs' are MRSA and *C. difficile*.

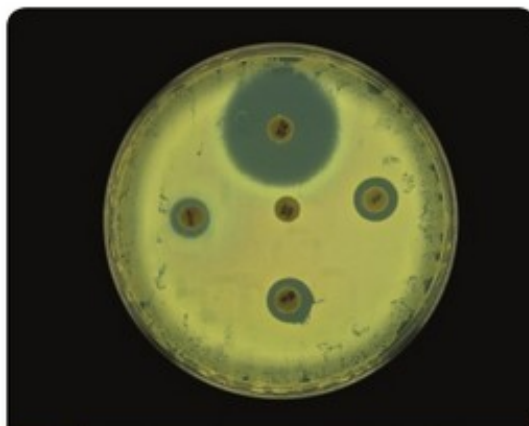
These bacteria are becoming widespread, especially in hospitals.



The antibiotic penicillin was first isolated in 1928 from a fungus by Sir Alexander Fleming.



12.2 The petri dish made famous by Alexander Fleming: penicillin fungus is at the top and the colonies of bacteria are at the bottom



12.3 A petri dish of bacteria with discs of different antibiotics: only one antibiotic is fully active against the bacteria

Potential abuse of antibiotics in medicine

- The overuse of antibiotics in medicine results in the increased growth of antibiotic-resistant bacteria (because they have no competition). This happens when doctors prescribe antibiotics unnecessarily (e.g. for virus infections). In some countries it is even legal to buy antibiotics over the counter (without a doctor's prescription).
- The failure of some patients to complete their treatment of antibiotics allows the bacteria to survive and re-grow. This leads to the need for more antibiotics (along with the increased risk of the growth of resistant bacteria).

DNA profiles

A **DNA profile** is a unique pattern of DNA from one person that is compared with the DNA profile of another person. DNA profiling is also called genetic or DNA fingerprinting.

Preparing DNA profiles

1 Release DNA from cells

DNA is released from cells in the same way as in the experiment to isolate DNA from plant tissue. The cells may be obtained from a saliva sample, the root of a hair, or a sample of semen.

2 Cut the DNA into fragments

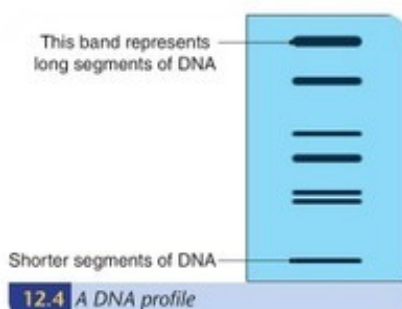
The DNA is cut into pieces using special enzymes (called restriction enzymes). These enzymes cut DNA when they encounter specific base sequences.

This is similar to cutting a section of text wherever a given word or sequence of words is found. The DNA sections obtained in this way will vary in length from very small sequences of bases to very long sequences.

3 Separate the fragments

The DNA fragments are separated according to their length. This involves placing the fragments in a gel and passing an electric current through the gel. Small fragments move faster through the gel than large ones. A photograph of the final results is obtained.

Each DNA profile looks like a bar code. The profile (or 'bar code') is different for each person. The likelihood of two people having the same DNA profile or fingerprint is very low (unless they are identical twins). A typical DNA profile is shown in the diagram.



Polymerase chain reaction (PCR)

Polymerase chain reaction (PCR) is a commonly used laboratory technique that produces multiple copies of a particular region of DNA which a researcher wishes to analyse or synthesize. DNA amplified by PCR may be sent for sequencing, visualized by a process known as gel electrophoresis, or cloned into a plasmid for further experiments.

The PCR process, like DNA replication in an organism, makes use of a polymerase enzyme that makes new strands of DNA, using existing strands as templates. Taq polymerase is the DNA polymerase typically used in PCR. It is named after the heat-resistant bacterium (*Thermus aquaticus*).

Once researchers have decided on the target area of DNA to be copied, they select primers which allow them to isolate it. A primer is a short piece of single stranded DNA which will bind to the opposite sequence in the DNA template. Two primers are used in PCR to bind to each end of the target region.

The Taq polymerase and primers are combined with the template DNA and nucleotides which are the building blocks of DNA and go through repeated cycles of heating and cooling that allow the target DNA to be synthesized.

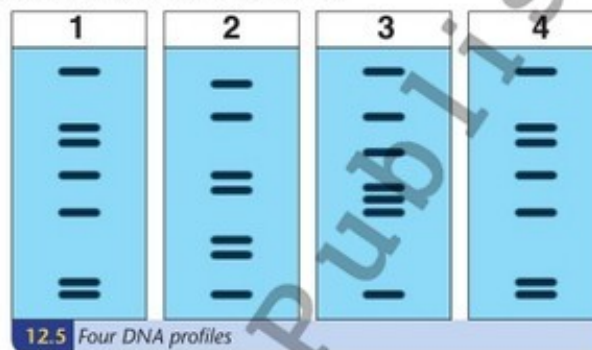
Uses of DNA profiles

Crime

DNA profiles may be used to link somebody to a crime or to the scene of a crime. If some biological tissue (such as saliva on a cigarette butt, a semen stain, or a hair) is found at the crime scene, its DNA profile is compared with one taken from a suspect. If the patterns match, the suspect is associated with the crime scene.

See, for example, the four DNA profiles below: the first is from the crime scene while the others are from suspects.

There is a match between number 1 and number 4.

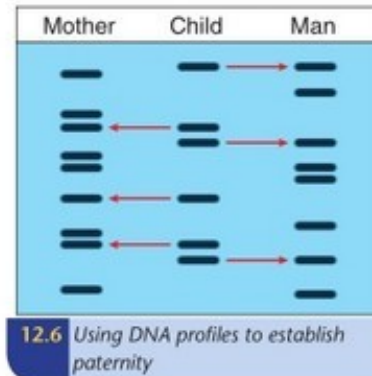


Medical

DNA profiles may be used to establish if a man is the father of a child (i.e. to establish paternity). Paternity cases are important in immigration, inheritance and rape cases. DNA profiles obtained from the mother, child and the man are compared.

If all of the child's bands match either one of the mother's or one of the man's bands, then the man is the father of the child, as shown below.

If some of the child's bands match the mother's but the others do not match the man's, then he is not the father of the child, as shown below.



Genetic screening

Genetic screening means testing a person's DNA for the presence of abnormal or altered DNA.

This process uses some of the techniques associated with DNA profiling.

The presence of abnormal or altered DNA is an indication that a particular gene is mutated.

Mutated genes are the cause of many genetic disorders such as:

- Cystic fibrosis (where fluid cannot be removed from the lungs)
- Haemochromatosis (where there is too much iron in the body)
- Albinism (the inability to form the pigment melanin)

The value of genetic screening

If a couple know that one or both of them have a particular mutation, they can be advised as to the probability that they will have a child affected by the disorder. They can then decide whether or not to have children.

If a child is born with a disorder, it is often helpful to be aware of this possibility in advance so that treatment can begin immediately.

Ethical problems

Genetic screening may cause problems, such as those listed below.

- If an embryo is tested and shown to have a disorder, it may encourage the couple to consider abortion.
- Should a person be told they have a disorder that will develop later in life and lead to death?
- Should insurance companies or potential employers be informed of genetic screening results?

Genetic engineering

The process of genetic engineering normally involves cutting a small section of DNA (usually containing a single gene called the target gene) from one organism and inserting it into the DNA of a second organism. In this respect it is really a 'cut and paste' process.

The altered DNA is called **recombinant DNA** because it recombines after the small section of DNA is inserted into it. The recombinant DNA is placed back into an organism.

The organism with the altered DNA is called a genetically modified organism (GMO). If this organism reproduces asexually, the altered DNA is also reproduced, so that all of the offspring get a copy of the recombinant DNA containing the target gene.

The organism with the altered DNA produces the substance for which the target gene codes, given the correct nutrients and conditions.

Genetic engineering is the artificial manipulation or alteration of genes.



12.7 A genetically engineered organism: it is half sheep and half goat

Alternative names for genetic engineering

Genetic engineering is known by a number of other names. These include genetic manipulation, genetic modification, recombinant DNA technology, genetic splicing ('to splice' means to join two overlapping strands together) and gene cloning.

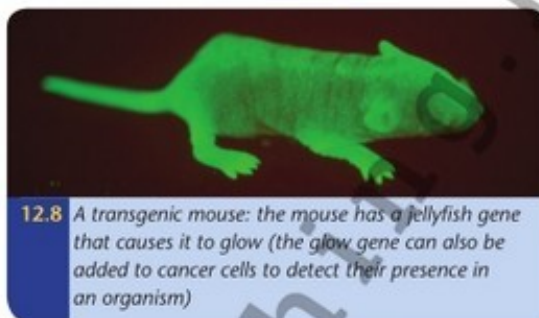
Gene cloning refers to the idea that many identical copies (clones) of the target gene are formed when the organism reproduces. Genetic engineering is the basis for most of the developments in the area of biotechnology.

Genetic engineering breaks the species barrier

Genetic engineering means that DNA from different species can be joined together. This often results in combinations of DNA that would never be possible in nature. This is because the two organisms would be prevented from reproducing together as they are from different species. For this reason, genetic engineering is not a natural process.

Examples of the cross-species transfer of genes are:

- Human genes can be inserted into bacteria
- Bacterial genes can be inserted into plants
- Human genes can be inserted into other animals.

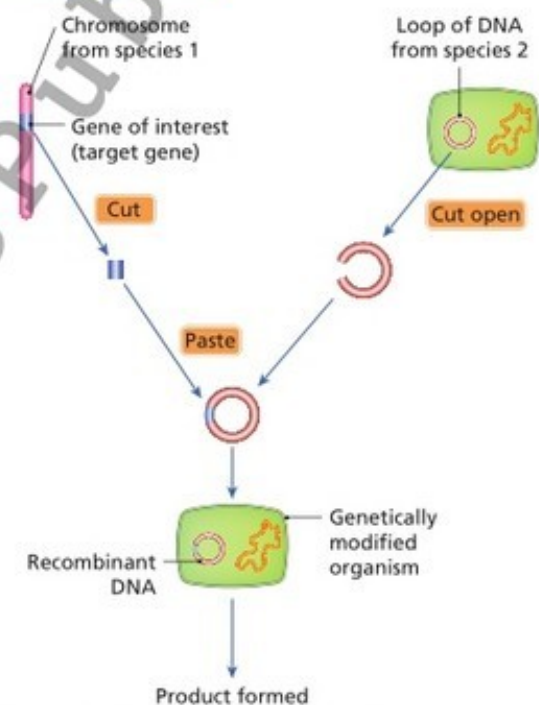


Substances used in genetic engineering

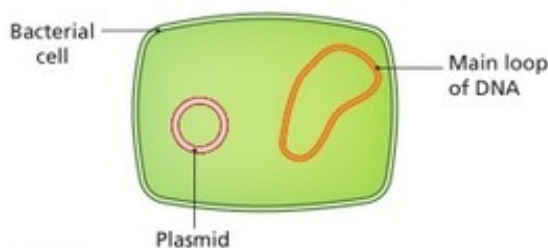
Certain tools and materials are needed in order to cut and paste pieces of paper. These include the original document, the new material that is to be pasted on to the original document, scissors and some glue or paste.

In much the same way, genetic engineering requires the following materials and tools:

- **A source of DNA.** This is the DNA (or the gene) that is taken from one organism to be placed into the DNA of a second organism. The inserted or target DNA can be thought of as 'foreign' DNA.
- **A cloning vector.** A cloning vector is a special kind of DNA that can accept foreign DNA and replicate (reproduce exactly) itself and the foreign DNA. The most common cloning vector is a bacterial plasmid. This is a small loop of DNA found in bacteria (in addition to the larger loop of DNA that acts as a bacterial chromosome).
- **Restriction enzymes.** These are enzymes that cut DNA at specific places. They act as the genetic 'scissors'.
- **DNA ligase.** DNA ligase is an enzyme that is used to combine the foreign DNA with the plasmid DNA. In this way DNA ligase acts as the genetic 'glue' or 'paste'.



12.9 Genetic engineering: a cut-and-paste process



12.10 DNA in a bacterium

Restriction enzymes

Restriction enzymes will cut DNA only at particular sites. Each restriction enzyme is specific to a particular sequence of DNA bases and will only recognise that specific sequence, cutting the DNA at those bases only. For example, one restriction enzyme will cut DNA whenever the base sequence GAATTC arises.

When DNA from two different organisms is cut using the same restriction enzyme, the cut ends from both sources will be complementary. If the cut sections are mixed together, they will form base pairs and combine to produce recombinant DNA.

DNA ligase

DNA ligase is an enzyme that is used to stick DNA molecules from different sources firmly together. In this case it acts as an anabolic enzyme. DNA ligase will only work if the DNA from the two sources has been cut with the same restriction enzyme. In this case, the ends of the cut DNA will be complementary to each other.

The process of genetic engineering

Genetic engineering is used in a vast and rapidly growing number of applications. The techniques used in most of these applications include the six steps outlined in the following sections. The examples will refer to a human gene being inserted into a bacterium. The human gene is the target gene.

Isolation

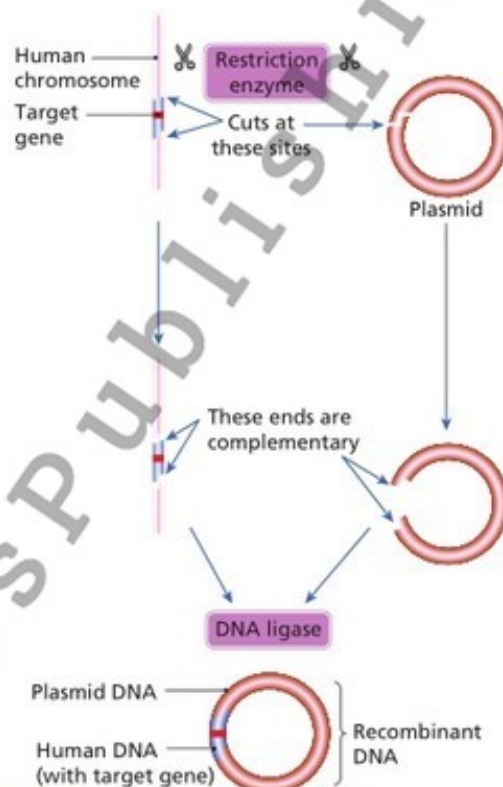
Isolation is the removal of the chromosome (containing the target gene) from the human cell and the plasmid DNA from the bacterium.

This is carried out in a similar manner to the way in which DNA is isolated from plant tissue.

Cutting

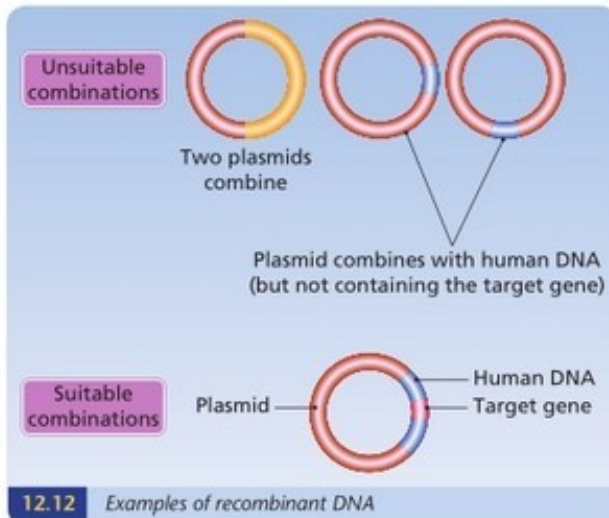
Normally the plasmid will be cut open at a single site. The human DNA (chromosome) will be cut into many sections. Only one of these sections will contain the target gene.

Diagram 12.11 shows a single plasmid and a single chromosome being cut. In reality large numbers of each are cut at this stage.



12.11 Producing recombinant DNA

The human DNA and the plasmid DNA are cut open using the same restriction enzyme.



Ligation

The cut plasmids are mixed with the human DNA sections. This allows the cut ends to combine. The intention is that the cut ends of the plasmid should combine (by base pairing) with the complementary ends of the human DNA that contains the target gene. This happens, but many other unsuitable combinations also form.

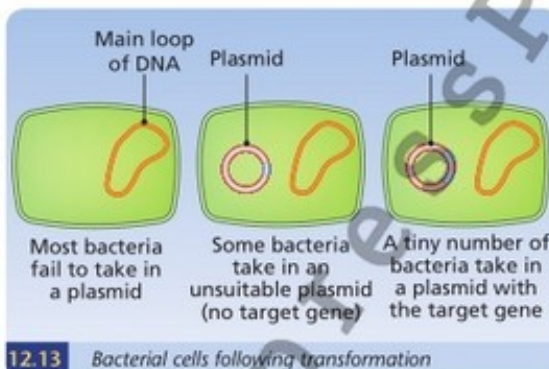
For example, the plasmids may stick to each other, a plasmid may stick to several human DNA sections in a

Ligation is the joining of two sections of DNA to form a single strand.

row, or a plasmid may stick to a section of human DNA that does not carry the target gene (see diagram 12.12).

Splicing is the joining of overlapping sections together. Ligation is the joining of the overlapping cut ends of the DNA sections. Ligation is also called DNA splicing.

DNA ligase is used to form the bonds within the recombinant DNA (i.e. between the plasmid DNA and the human DNA). In this way the target gene becomes part of the plasmid DNA.



Transformation

Transformation is the uptake of DNA into a cell.

The bacteria are treated in such a way that they can take in plasmids (composed of recombinant DNA) from a surrounding solution.

The vast majority (up to 99%) of the bacterial cells normally fail to take up the

plasmids or only take up a plasmid that does not contain the target gene. These bacteria will be of no further use because they do not contain the target gene. Special techniques are used to identify the small number of bacteria that have taken up a plasmid containing the target gene (i.e. the bacteria containing the recombinant DNA).

Cloning

Cloning is the production of identical copies of the bacterium (containing the target gene).

The bacteria containing the target gene are grown (or cloned) using a nutrient medium. As the bacteria reproduce, they produce copies of the plasmid with the target gene.

Expression

Expression is the formation of the product by the organism with the recombinant DNA.

Expression normally takes place in a bioreactor. Once the product has been formed (or expressed) in sufficient amounts, it has to be separated from the culture and the bacteria that produced it.



12.14 Spraying weed-killer on herbicide-resistant soya plants

Applications of genetic engineering

Plants

Weed-killer-resistant crops

Many crop plants have bacterial genes added to them. These plants are then resistant to particular weed-killers (or herbicides). This means that when the herbicide is sprayed on the crop it will kill the weeds but will not kill the transgenic plants.

Animals

Sheep produce a protein to treat emphysema

Some people have a faulty gene that means they cannot produce a protective protein in their lungs. This leads to the collapse of the alveoli, in a condition known as emphysema.

A working human gene for this protein (known as AAT) has been inserted into sheep DNA. The sheep can then produce the protein in their milk.



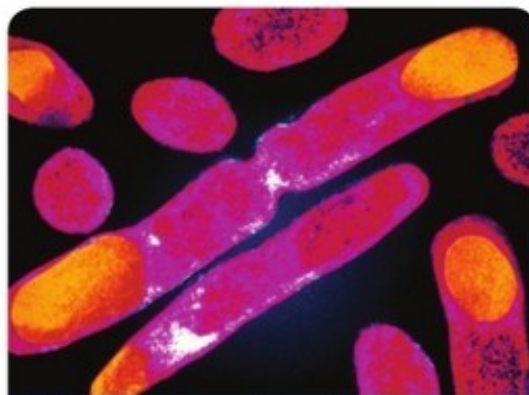
12.15 Transgenic sheep: these sheep produce a protein to treat emphysema

Microorganisms

Bacteria make insulin

One of the first genetically engineered proteins to be produced commercially was human insulin. This involved inserting the gene for human insulin into a bacterium (called *E. coli*). The bacterium then produced large quantities of insulin. This meant that people with diabetes (who have a shortage of insulin) could inject themselves with human insulin.

Prior to this development insulin was obtained from animals. This caused problems because many diabetics are allergic to animal insulin.



12.16 Insulin-producing bacteria: insulin is made in the orange parts

Advantages and disadvantages of using living organisms in biotechnology

The goals of biotechnology include the making of improved products, the improvement of crops and livestock and their protection against different forms of disease, the development of microorganisms for a range of specific applications in agriculture and the development of a range of treatments and pharmaceuticals for human health.

Advantages

There are a range of potential economic, environmental and medical research benefits relating to the uses of biotechnology which include:

- creating greater growth rates in livestock or crop yields
- creating greater disease resistance in herds or crops
- improved use of dietary phosphorous to reduce the environmental impact of manure
- enhancing nutritional benefits for humans of animal products such as meat eggs and milk
- the use of organs grown in animals in human transplant procedures
- the development of new treatments for human disease

Disadvantages

These relate to three specific areas: environmental concerns, food safety and social concerns connected with animal welfare and consumer rights.

Environmental concerns

Changing animal feed sources and introducing transgenic animals into existing animal populations can upset the ecological balance, by affecting patterns of predation and disruption of reproduction patterns.

Food safety

There is concern that:

- biotechnology may generate new allergens through the introduction of new proteins into organisms
- there can be unforeseen consequences of introducing functional proteins such as growth hormone to animals for humans that consume the animal as food
- the toxicity of unintended expression products and the changing of the nutritional profile of a product.

Animal welfare and social concerns

There are a wide range of these but most commonly these relate to:

- people condemning research and farming practices implicated in or resulting from biotechnology that may be considered cruel or unnatural
- how biotechnologies may affect traditional or seasonal human interactions with the natural environment
- the labelling of products as being generated from genetic-engineering technology which would then give people the opportunity to reject them.

In small groups research specific examples of the uses and effects of biotechnology which relate to specific advantages and disadvantages above, and then prepare a short group presentation for class on one specific use of biotechnology that clearly demonstrates one of its advantages and one specific use which highlights one of its disadvantages.

Ethical issues in genetic engineering

The use of genetically modified organisms provides undoubted benefits. Nevertheless, these techniques raise safety and ethical issues for people and the environment. These issues centre on concerns such as:

- The release of GMOs into the environment
- The use of GMOs as a food source
- The concern that animals will suffer as a result of being genetically modified
- The fear that humans (especially human zygotes) may be genetically modified.



12.17 *A genetically modified pig: the pig has human genes so that it produces organs for transplant into humans*

Biotechnology in Kazakhstan

The National Centre for Biotechnology in Astana is Kazakhstan's largest scientific centre, indicating the importance of this area of science to the country's future development. Research in the field of biotechnology is important across many sectors of national life. Key areas include: human and animal health, development of agriculture, food safety and preservation and environmental areas such as waste water treatment and the treatment of industrial waste. The newspaper article below discusses one such area of innovation in the country that has led to the development of a range of dental care products. The extract from a journal points to promising research in improving wheat yields in Kazakhstan. Read the article and extract and then discuss in groups the innovative use of biotechnology behind the development of the product/research.



Kazakh biotechnologist aims to help billions live without tooth decay

October 2018

Reported recently in the Astana Times is the news that Kazakh biotechnologist Bauyrzhan Aituov has produced an innovative new product for treating tooth decay, Innodent, which avoids the need for drilling and fillings. According to Aituov, the product prevents early tooth decay by stimulating natural regeneration of tooth enamel.

The product Innodent Repair is based on a synthesised human protein called amelogenin. This protein is present in fetal enamel development but not in fully developed teeth. With no substitute protein available in nature, the protein is synthesised in laboratories. Innodent repair is applied to enamel damaged by tooth decay and the protein interacts with minerals in saliva [phosphorus, calcium and magnesium] and thereby regenerates tooth enamel and repairs the damage. According to Aituov, the advantage over traditional methods of treatment such as drilling and filling is that Innodent Repair is a regenerative cure for tooth decay. He points to nature as the source of the inspiration for the innovation, noting that animals such as sharks do not suffer from tooth decay because of the thickness of their enamel.

Kazakhstan's National Agency for Technological Development initially supported the company with a grant to purchase automated equipment and the product was first produced in 2016.



A recent report in the Journal of Biotechnology and Biomaterials points to promising research by Galiya Akhmetova from the A I Barayev Scientific Production Centre for Grain Farming, in using a biological preparation of the bacteria *Pseudomonas* to improve wheat seed energy growth and seed germination in wheat varieties in Northern Kazakhstan. In tests, wheat inoculated with strains of *Pseudomonas* exhibited improved growth when compared to control samples.

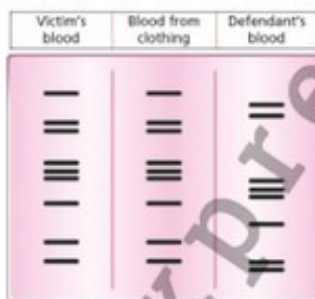
In small groups research another innovative area of research or product development in the field of biotechnology in Kazakhstan and prepare a short slide presentation for the rest of class.

Your presentation should cover:

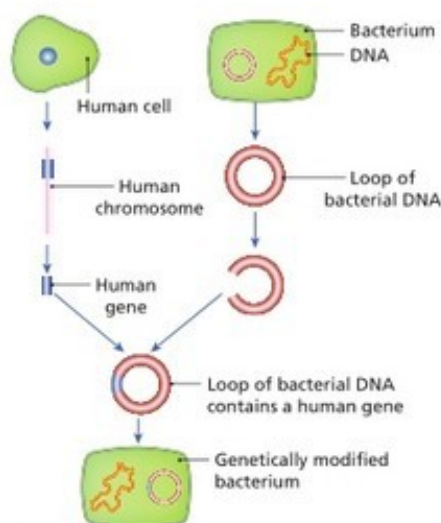
- Background and Kazakhstan context of the research/product
- Biology and biotechnology involved
- Impact or potential impact of the innovation
- Possible limitations of the research/product

Questions on Module 12

- 1 Name two types of microorganisms used in technology.
- 2 Give (a) two industrial uses and (b) two medical uses of biotechnology.
- 3 Suggest a biological explanation for the following:
Doctors are reluctant to prescribe antibiotics to patients suffering from common cold-like symptoms.
- 4 (a) What is genetic profiling?
(b) Give an outline account of how a genetic profile (or 'fingerprint') is obtained.
(c) On a genetic profile, what does each band represent?
(d) Explain why some of the bands are thicker than others.
- 5 (a) Give two applications for genetic profiling.
(b) After a violent crime, genetic profiles were carried out on the victim's blood, the defendant's blood and some blood stains obtained from the clothing of the defendant. The results are given in the diagram below. What conclusions can be drawn from these results?



- 6 (a) Name one human condition that may be identified through genetic screening.
(b) State one possible benefit and one problem associated with genetic screening.
- 7 Explain what is meant by the following terms:
 - (a) Genetic engineering
 - (b) Target gene
 - (c) Recombinant DNA
 - (d) Splicing
 - (e) Transgenic.
- 8 Suggest one reason why genetic engineering is not a natural process.
- 9 Give one use for each of the following:
 - (a) A cloning vector
 - (b) A restriction enzyme
 - (c) DNA ligase
 - (d) A plasmid.
- 10 Explain the meaning of each of the following terms in relation to forming a genetically modified bacterium containing the gene for human growth hormone (HGH):
 - (a) Isolation
 - (b) Cutting
 - (c) Ligation
 - (d) Transformation
 - (e) Expression.
- 11 Explain, with the aid of a diagram, why genetic engineering can be considered to be a 'cut-and-paste' process.
- 12 Give one example and state a benefit for each of the following:
 - (a) Inserting a human gene into a bacterium
 - (b) Inserting a bacterial gene into a plant
 - (c) Inserting a human gene into an animal.
- 13 Human growth hormone (HGH) is produced in the pituitary gland. People who do not produce sufficient amounts of HGH do not grow properly. The gene for HGH can be extracted from a human chromosome and inserted into a loop of DNA in a bacterium, as shown in diagram 12.18. This procedure allows the hormone to be produced by bacteria using genetic engineering techniques.


12.18 Producing HGH in a bacterium

- What is a hormone?
 - Where in the body is the pituitary gland located?
 - What are chromosomes made of?
 - What biomolecules are used to extract a gene from a chromosome?
 - Explain why the same biomolecule must be used to extract the gene and to open the loop of DNA in the bacterium.
 - What substances should be added to a bioreactor to enable bacteria to grow?
- Choose which of the options (i), (ii), (iii) or (iv) represents the correct answer in each case below.
 - Genetic engineering:
 - Is a natural process
 - Only takes place in microorganisms
 - Happens when cells divide
 - Involves combining DNA from different species.
 - The most common cloning vector used in genetic engineering is:
 - RNA
 - An enzyme
 - A plasmid
 - DNA ligase.
 - Which of the following is least associated with genetic engineering?
 - Translation
 - Transformation
 - Cloning
 - Expression.
 - Genetically modified organisms:
 - Are always harmful
 - Are always microorganisms
 - May be beneficial
 - Arise naturally.
 - Some antibiotics, like penicillin, are naturally produced by fungi that live in soils where there are not many nutrients. Suggest why some fungi that live in soil produce substances to kill bacteria.
 - Antibiotics have different ways of working. Some prevent bacterial cell wall formation and others prevent ribosomes from working. Use these facts to explain why antibiotics do not work on viruses.
 - The antibiotic tetracycline acts by attaching to bacterial ribosomes and preventing the ribosomes from working.
 - Predict the effect this will have on bacteria.
 - Tetracycline binds to bacterial ribosomes but **not** human ribosomes. What does this suggest about the ribosomes in bacteria and humans?
 - When fruit juice is produced, it can be cloudy due to the presence of pectin which is an insoluble polysaccharide found between plant cells. The enzyme pectinase can be used to clear the fruit juice as it breaks down pectin.
 - Suggest what the product of the pectinase reaction is **and** why this clears the fruit juice.
 - Suggest why only the pectinase enzyme is used and **not** a whole organism, such as the fungus which makes pectinase.
 - When DNA fragments are being separated in a gel, they move toward the positive terminal. What does this suggest about the DNA molecule?

Glossary

A

abiotic factors non-living factors.

acquired variations are not inherited but are learned or developed during life.

active immunity the production of a person's own antibodies in response to foreign antigens that enter the body.

active site the part of an enzyme that combines with the substrate.

active transport energy (in the form of ATP) is used to move molecules, often against a concentration gradient, i.e. from low concentrations to high concentrations.

adaptation any alteration that improves an organism's chances of survival and reproduction

adhesion occurs when different molecules stick together.

aerobic respiration the controlled release of energy from food using oxygen.

alleles different (or alternative) forms of the same gene.

all or nothing law states that if the threshold is reached an impulse is carried, but if the threshold is not reached no impulse is carried.

amplification (DNA) An increase in the frequency of a gene or chromosomal region, for example, by gene duplication or polymerase chain reaction.

anabolic reactions use energy to convert smaller molecules into larger molecules.

anaerobic respiration the controlled release of energy from food without the use of oxygen.

antagonistic pair two muscles that have opposite effects to each other.

antibiotics chemicals produced by microorganisms that stop the growth of, or kill, other microorganisms without damaging human tissue.

antibody a protein produced by white blood cells (called lymphocytes) in response to a specific antigen.

anticodon a sequence of three bases (a triplet) on tRNA that are complementary to a sequence of three bases on mRNA.

antigen a foreign molecule that stimulates the production of antibodies.

appendicular skeleton composed of the limbs (arms and legs), the pectoral (shoulder) girdle and the pelvic (hip) girdle.

artificial active immunity occurs when a pathogen is medically introduced into the body.

artificial passive immunity occurs when a person is given an injection containing antibodies made by another organism.

aseptic or **asepsis** means that measures are taken to exclude unwanted microorganisms.

asexual reproduction involves only one parent.

autotrophic (organism) one that makes its own food.

axial skeleton consists of the skull, spine, ribs and sternum (breastbone).

B

bacteriophage (or **phage**) a virus that infects bacteria.

balanced diet contains all the necessary food types in the correct proportions.

batch culture the growth of cells in a sealed container (or bioreactor) over a short period of time and under ideal conditions until all the nutrients are used up.

biogenesis living things arise from other living things of the same type. This is also called **continuity of life**.

biology the study of living things.

biomolecules chemicals that are made inside a living thing.

bioprocessing the use of enzyme-controlled reactions to produce a product.

bioreactor a vessel or container in which living cells or their products are used to make a product.

biosphere that part of the planet containing living organisms.

biotechnology the use of living things or their components (especially cells and enzymes) to manufacture useful products or to carry out useful reactions.

biotic factors living factors.

birth control methods taken to limit the number of children that are born.

blastocyst (or **blastula**) a hollow ball of cells formed from a morula.

blood pressure the force exerted by the blood against the walls of the blood vessels (mainly the arteries).

bud a potential growth point that may develop into a shoot, a leaf or a flower.

bulb a modified bud.

C

cancer a group of disorders in which certain cells lose their ability to control both the rate of mitosis and the number of times mitosis takes place.

carnivores animals that feed mainly on animals. Examples are dogs, cats and ladybirds.

carpels the female parts of the flower.

catabolic reactions release energy when a complex molecule is broken down to a simpler form.

catalyst a substance that speeds up a reaction, without itself being used up in the reaction.

cell continuity all cells develop from pre-existing cells.

cell cycle the changes that take place in a cell during the period between one cell division and the next.

cellular energy the energy stored in the bonds of biomolecules.

centromere the point at which the chromosomes are attached in a double-stranded chromosome.

characteristics traits or features that are inherited genetically.

chemosynthesis the production of food using energy released from chemical reactions.

chemotropism a change in growth of a plant in response to chemicals.

chromatin the name given to chromosomes when they are elongated and not dividing.

chromosomes coiled threads of DNA (which forms genes) and protein that become visible in the nucleus at cell division.

chromosome mutation a large change in the structure or number of one or more chromosomes.

classification placing objects into groups based on similar characteristics.

climatic factors refer to weather over a long period of time.

cloning the production of identical copies of the bacterium (containing the target gene).

closed circulatory system blood remains in a continuous system of blood vessels.

codominance neither allele is dominant or recessive with respect to the other. Both alleles are equally expressed in the heterozygous genotype to produce an intermediate phenotype.

codon (or **triplet**) a sequence of three bases in DNA (or RNA) that act as a code for an amino acid.

cohesion the sticking of similar molecules to each other.

community all the different populations in an area.

competition occurs when organisms actively struggle for a resource that is in short supply.

conclusion a summary of the results of an experiment.

conservation the wise management of the existing natural resources in an ecosystem, in order to maintain a wide range of habitats and prevent the death and extinction of organisms.

consumers organisms that take in food from another organism.

contest competition an active physical contest between two individual organisms.

continuity of life living things arise from other living things of the same type. This is also called **biogenesis**.

continuous flow (food processing) the growth of cells in an open container (or bioreactor), where nutrients are added and the end products are removed all the time at a rate that maintains the volume of liquid and the number of cells.

contraception the deliberate prevention of fertilisation or pregnancy.

control used to provide a comparison (or standard) against which the actual experiment can be judged.

copulation the act of sexual intercourse.

cotyledon a seed leaf.

cross-pollination the transfer of pollen from an anther to a stigma on a different plant.

cutting a portion of a plant that is removed from the parent plant and grown into a new, independent plant.

cytoplasm the living material in a cell outside the nucleus.

D

data the measurements, observations or information gathered from experiments.

decomposers organisms that feed on dead organic matter.

denatured enzyme one that has lost its shape and can no longer carry out its function.

denitrification the conversion of nitrates to nitrogen gas.

detritus feeders organisms that feed on small pieces of dead organic matter.

diastole when the heart chambers relax.

diffusion the spreading out of molecules from a region of high concentration to a region of low concentration.

dihybrid cross involves the study of two characteristics.

diploid cell one that has two sets of chromosomes, i.e. it has two of each type of chromosome in the nucleus.

dispersal the transfer of a seed or fruit away from the parent plant.

DNA profile (also called a DNA or genetic fingerprint) a method of making a unique pattern of bands from the DNA of a person, which can then be used to compare with the DNA profile of another person.

dominant the allele that prevents the recessive allele from being expressed.

dormancy a resting period when seeds undergo no growth and have reduced cell activity or metabolism.

double blind both the investigator and the participant are unaware of the nature of the treatment the participant is receiving.

E

ecological niche (of an organism) the functional role it plays in the community.

ecology the study of the interactions between living things (organisms) and between organisms and their environment.

ecosystem a group of clearly distinguished organisms that interact with their environment as a unit.

ectotherms gain or lose heat from or to their external environment.

edaphic factors relate to soil.

ejaculation the release of semen from the penis.

endocrine gland a ductless gland that produces hormones, which are released directly into the bloodstream.

endospermic seed contains some endosperm when fully formed.

endotherms generate their own heat from metabolic reactions.

enzymes proteins that speed up a reaction without being used up in the reaction.

enzyme specificity each enzyme will react with only one particular substrate.

ethics relates to whether conduct is right or wrong.

eukaryotic cells have a nucleus and cell organelles, all of which are enclosed by membranes.

evolution the way in which living things change genetically to produce new forms of life over long periods of time.

excretion the removal of waste products of metabolism from the body.

exhalation breathing out.

exocrine glands release their product into ducts or tubes.

experiment a test for a hypothesis.

expression the formation of the product by the organism with the recombinant DNA.

F

facultative parasite can get its food from a live or a dead host.

fauna all the animals in an ecosystem.

fermentation another name for anaerobic respiration.

fertilisation the union of the male and female gametes to form a diploid zygote.

filtration water and small molecules pass (under high pressure) from the blood into the nephron.

flora all the plants in an ecosystem.

food chain (grazing food chain) a sequence of organisms in which each one is eaten by the next member in the chain.

food web two or more interlinked food chains.

forensic medicine the way in which medical knowledge is used in legal situations.

fossil the remains of something that lived a long time ago (or some indication of something that lived a long time ago).

frequency the chance of finding a named species with any one throw of a quadrat.

fruit a developed ovary.

G

gametes haploid cells capable of fusion.

ganglion (plural: ganglia) a group of cell bodies located outside the CNS.

gene a section of DNA that contains the instructions for the formation of a protein.

gene expression the way in which the genetic information in a gene is decoded in the cell and used to make a protein.

gene (or point) mutation a change in a single gene.

general defence system acts as a barrier to all pathogens attempting to gain entry to the human body.

genetic code the sequence of bases in DNA that provide the instruction for a cell (using RNA) to form a protein.

genetic engineering the artificial manipulation or alteration of genes.

genetic screening testing DNA for the presence or absence of a particular gene or an altered gene.

genotype the genetic make-up of an organism, i.e. the genes that are present.

geotropism (or gravitropism) the change in growth of a plant in response to gravity.

germination the regrowth of the embryo, after a period of dormancy, if the environmental conditions are suitable.

germ layers basic layers of cells in the blastocyst from which all adult tissues and organs will form.

gestation the length of time spent in the uterus from fertilisation to birth.

glycolysis the conversion of glucose into two molecules of pyruvic acid.

gonad an organ that produces sex cells in animals.

grafting the joining and uniting of part of one plant with a second plant.

growth inhibitor a chemical that causes a reduction in growth of plants.

growth plate the area between the epiphysis and the diaphysis in a long bone within which bone growth occurs.

growth promoter a chemical that causes increased growth in plants.

growth regulator a chemical that controls the growth of a plant.

H

habitat the place where a plant or an animal lives (and is also the local area of study).

haploid cell one that has one set of chromosomes, i.e. it has only one of each type of chromosome in the nucleus.

helper T cells stimulate B cells and killer T cells.

herbaceous plants do not contain wood (or lignin).

herbivores animals that feed mainly on plants. Examples are sheep, cattle and rabbits.

heredity the passing on of features from parents to offspring by means of genes.

heterotrophic (organism) one that takes in food made by other organisms.

heterozygous the alleles are different.

homeostasis the ability of an organism to maintain a constant internal environment.

homologous pair two chromosomes of similar size with the same sequence of genes.

homozygous two alleles that are identical.

hormone a chemical messenger produced by an endocrine gland and carried by the bloodstream to another part of the body, where it has a specific effect.

hydrotropism a change in growth of a plant in response to water.

hypha a tube or filament in a fungus.

hypothesis an educated guess based on observations.

I

immobilised enzymes are attached, or fixed, to each other, or to an inert material.

immunisation occurs when we produce or are injected with antibodies against a pathogen.

immunity the ability to resist infection.

implantation the embedding of the fertilised egg into the lining of the uterus.

induced immunity the ability to resist disease caused by specific pathogens by the production of antibodies.

infertility the inability to produce offspring.

inhalation breathing in.

inherited variations are controlled by genes.

insemination the release of semen into the vagina, just outside the cervix.

interneuron (also called an **intermediate, relay** or **association neuron**) carries information between sensory and motor neurons.

internode the region on a stem between two nodes.

interphase the phase in the cell cycle when the cell is not dividing.

inter-specific competition occurs between members of different species.

intra-specific competition occurs between members of the same species.

in-vitro fertilisation (IVF) removing eggs from an ovary and fertilising them outside the body.

isolation the removal of the chromosome (containing the target gene) from the human cell and the plasmid DNA from the bacterium.

J

joint where two or more bones meet.

K

key a means of naming organisms by answering a series of questions with alternative answers.

killer T cells destroy abnormal human body cells.

L

lactation the secretion of milk by the mammary glands (breasts) of the female.

larynx the voice box.

law or **principle** arises from a theory that has been shown to be valid when fully tested over a long period of time.

law of independent assortment states that: when gametes are formed either of a pair of alleles is equally likely to combine with either of another pair of alleles.

law of segregation (Mendel's first law) states that:

- Inherited characteristics are controlled by pairs of alleles.
- These alleles segregate (or separate) from each other at gamete formation, with only one member of the pair being found in each gamete.

layering the growth of a new plant from a stem that is still attached to the parent plant.

lenticels openings in the stems of plants that allow gas exchange.

life the possession of all the following characteristics: organised, requiring nutrition and excretion, capable of responding and reproducing.

ligaments strong, fibrous, slightly elastic tissues that connect bone to bone.

ligation the joining of two sections of DNA to form a single strand.

lignin a strengthening material found in some plant cell walls.

linkage genes are located on the same chromosome.

locus (of a gene) its position on a chromosome.

M

meiosis a form of nuclear division in which the four daughter nuclei contain half the chromosome number of the parent nucleus.

memory B cells survive for years after the infection is eliminated and can make the specific antibody if the same infection later enters the body.

memory T cells survive for years after the infection is eliminated and can stimulate the specific B cells and killer T cells if the same infection later enters the body.

menopause when ovulation and menstruation stop happening in a female.

menstrual cycle a series of events that occurs every 28 days on average in the female if fertilisation has not taken place.

menstruation the discharge of the lining of the uterus (the endometrium) and the unfertilised egg.

meristem a plant tissue capable of mitosis.

metabolism the sum of all the chemical reactions in an organism.

microorganisms small living things.

micropropagation the growth of plants from small pieces of tissue under sterile conditions on a specially selected medium.

mitosis a form of nuclear division in which one nucleus divides to form two nuclei, each containing the same number of chromosomes with identical genes.

monohybrid cross involves the study of a single characteristic.

morula a solid ball of cells formed from a zygote by mitosis.

motor (or **efferent**) **neuron** takes a message from the CNS to a muscle or a gland.

mutagens agents that cause mutations.

mutation a spontaneous (or sudden) change in the amount or structure of DNA.

mycelium a (usually) visible mass of hyphae.

N

natural active immunity occurs when a pathogen enters the body in the normal way (i.e. when you get an infection).

natural passive immunity occurs when a child gets antibodies from its mother.

natural selection the process by which those organisms with genetically controlled characteristics that allow them to be well adapted to their environments will survive and reproduce to pass on their genes to following generations.

natural vegetative propagation involves forming new plants from a stem, root, leaf or bud.

negative tropism occurs when the growth is away from the stimulus.

neuron (or **neurone**) a nerve cell.

nitrification the conversion of ammonia and ammonium (NH_4^+) compounds to nitrite and then to nitrate.

nitrogen fixation the conversion of nitrogen gas into ammonia (NH_3), ammonium (NH_4^+) or nitrate (NO_3^-).

node the point on a stem at which a leaf is attached.

non-endospermic seed has no endosperm when fully formed.

nutrient recycling the way in which elements (such as carbon and nitrogen) are exchanged between the living and non-living components of an ecosystem.

nutrition the way organisms obtain and use food.

O

obligate parasite can only take its food from a live host.

observation when something is noticed.

omnivores animals that feed on plants and animals. Examples are humans, badgers and hedgehogs.

open circulatory system blood leaves blood vessels and flows around the cells of the animal's body before re-entering blood vessels again.

optimum pH the pH value at which the enzyme works best.

organ a structure composed of a number of tissues that work together to carry out one or more functions.

organisation living things are composed of cells, tissues, organs and organ systems.

organism a living thing.

organ system a number of organs working together to carry out one or more functions.

orgasm the physical and emotional sensations experienced at the peak of sexual excitement.

osmosis the movement of water molecules across a semi-permeable membrane from a region of high water concentration to a region of low water concentration.

osteoblast a bone-forming cell.

osteoclast a bone-digesting cell.

ovulation the release of an egg from the ovary.

P

parasites organisms that take in food from a live host and usually cause harm.

parasitism when two organisms of different species live in close association and one organism (the parasite) obtains its food from, and to the disadvantage of, the second organism (the host).

passive immunity occurs when individuals are given antibodies that were formed by another organism.

pathogen an organism that causes disease.

pathogenic bacteria bacteria that cause disease.

pectoral girdle consists of the collarbone (or clavicle) and the shoulder blade (or scapula).

pedigree a diagram showing the genetic history of a group of related individuals.

pelvic girdle composed of two halves of the hip joined to the sacrum.

percentage cover an estimate of the amount of ground in a quadrat covered by each species.

peristalsis a wave of muscular action in the walls of the alimentary canal that moves the contents along.

pharynx the throat.

phenotype the physical make-up, or appearance, of an organism.

phospholipids fat-like substances in which one of the fatty acids is replaced by a phosphate group or has a phosphate group added to it.

photolysis the splitting of water by light.

phototropism the change in growth of a plant in response to light, usually from one direction (i.e. unidirectional light).

plasma the liquid part of blood.

plasma B cells produce antibodies.

plumule the part of the plant embryo that develops into the shoot.

pollination the transfer of pollen from an anther to a stigma of a flower from the same species.

pollutants harmful additions to the environment.

pollution any harmful addition to the environment.

population all the members of the same species living in an area.

portal system a blood pathway that begins and ends in capillaries.

positive tropism occurs when the growth is towards the stimulus.

predation the catching, killing and eating of another organism.

predator an organism that catches, kills and eats another organism.

prey the organism that is eaten by a predator.

principle (or law) arises from a theory that has been shown to be valid when fully tested over a long period of time.

producers organisms that carry out photosynthesis.

product the substance(s) formed by an enzyme.

progeny offspring that are produced.

prokaryotic cells do not have a nucleus or membrane-enclosed organelles.

protoplasm all the living parts of a cell.

puberty the beginning of sexual maturity.

pulse the alternate expansion and contraction of the arteries.

punnett square a grid used to show the ratio of the genotypes of the progeny in a genetic cross.

purines (double-ring molecules) adenine (A) and guanine (G).

pyramid of numbers represents the numbers of organisms at each trophic level (or stage) in a food chain.

pyrimidines (single-ring molecules) thymine (T) and cytosine (C).

Q

qualitative study records the presence or absence of organisms.

R

radicle the part of the plant embryo that develops into a root.

reabsorption molecules pass from the nephron back into the blood.

recessive the allele is prevented from being expressed by a dominant allele.

reflex action an automatic, involuntary, unthinking response to a stimulus.

reflex arc the pathway taken by a nerve impulse in a reflex action.

refractory period a short time span after a neuron has carried an impulse during which a stimulus fails to cause a response.

replicate a repeat of an experiment.

reproduction the production of new individuals.

response the activity of a cell or organism as a result of a stimulus.

root tuber a swollen, underground root that remains dormant during winter and from which new plants may grow.

runners horizontal stems that run (or grow) above ground and from which new plants grow.

S

saprophytes organisms that take in food from dead organic matter.

scientific method a process of investigation in which problems are identified and their suggested explanations are tested by carrying out experiments.

scramble competition all of the competing individuals get some of the resource.

secondary sexual characteristics those features that distinguish males from females, apart from the sex organs themselves.

secretion some substances pass from the blood into the nephron.

selectively permeable membrane allows some but not all molecules to pass through.

self-pollination the transfer of pollen from an anther to a stigma on the same plant.

semen a fluid containing sperm and seminal fluid.

sensory (or afferent) neuron takes a message from a sense organ to the CNS.

serum plasma from which the clotting proteins have been removed.

sex linkage a characteristic is controlled by a gene on a sex (or X) chromosome.

sexual reproduction the union of two sex cells or gametes.

solar energy energy from the Sun.

speciation the production of new species as a result of evolution.

species a group of similar organisms that are capable of naturally interbreeding with each other to produce fertile offspring.

specific defence system attacks particular (or specific) pathogens.

sporulation the process of making spores.

stamens the male parts of the flower.

sterile all microorganisms are destroyed, i.e. there is nothing living.

stimulus (plural: stimuli) anything that causes a reaction in an organism or in any of its parts.

substrate the substance with which an enzyme reacts.

suppressor T cells inhibit the immune response.

symbiosis occurs when two organisms of different species live (and have to live) in close association and at least one of them benefits.

synapse a region where two neurons come into close contact.

synaptic cleft the tiny gap between the two neurons at a synapse.

systole when the heart chambers contract.

T

taxonomy the science of classifying organisms.

tendons strong, flexible, inelastic fibres that connect muscle to bone.

theory a hypothesis that has been supported by many different experiments.

thigmotropism a change in growth of a plant in response to touch.

threshold the minimum stimulus needed to cause an impulse to be carried in a neuron.

tissue culture the growth of cells in or on a sterile nutrient medium outside an organism.

tissue a group of similar cells that are modified (or adapted) to carry out the same function(s).

transcription the copying of a sequence of genetic bases from DNA onto messenger RNA (mRNA).

transformation the uptake of DNA into a cell.

translation the conversion of a sequence of genetic bases on messenger RNA into a sequence of amino acids.

transpiration the loss (by evaporation) of water vapour from the leaves and other aerial parts of a plant.

triplet (or **codon**) a sequence of three bases in DNA (or RNA) that act as a code for an amino acid.

trophic level a feeding stage in a food chain.

tropism a change in the growth of a plant in response to an external stimulus.

turgor (or **turgor pressure**) the outward pressure of the cytoplasm and vacuole against the cell wall of a plant.

U

ultrastructure the detail of a structure as seen using an electron microscope.

V

vaccination the administration (usually by injection) of a non-disease-causing dose of a pathogen (or its toxin) to stimulate the production of antibodies.

vaccine a non disease-causing dose of a pathogen (or its toxin), which triggers the production of antibodies.

valves control the direction of blood flow.

variable a factor that may change in an experiment.

variation (within a species) in a group of successfully interbreeding organisms the individual members show different characteristics.

vegetative propagation (or **vegetative reproduction**) asexual reproduction in plants.

venation the pattern of veins in a leaf.

woody plants contain wood (or lignin).

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