

GCSE 9-1 BIOLOGY

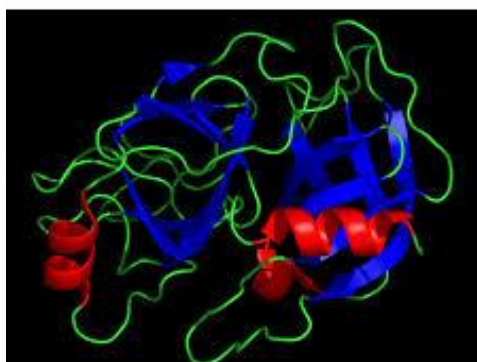


Paper 1

Revision booklet

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Topic 1 – Key concepts in Biology

Revision booklet

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Microscopy

Extremely small structures such as cells cannot be seen without microscopes, which enlarge the image.

Light Microscopes

The most commonly used microscope is the light **microscope**. This was invented by Robert Hooke in the 1600's.

- It has two lenses (**eyepiece lens and objective lens**)
- It is usually illuminated from underneath
- Used to view tissues, cells and large sub-cellular structures

To work out a microscope's magnification you multiply the magnification of the eyepiece lens and the objective lens. So if the magnification of a microscope with a x10 eyepiece lens and x20 objective lens is $10 \times 20 = \times 200$.

Electron Microscopes

In the 1930s the electron **microscope** was developed, enabling scientists to view deep inside sub-cellular structures, such as mitochondria, ribosomes, chloroplasts and plasmids. Electrons pass through a sample to build an image. They have a greater magnification power and a greater resolution.

There are two types: a scanning **electron microscope** that create 3D images (at a slightly lower magnification) and a **transmission electron microscope** which creates 2D images detailing organelles

Comparing the two types of microscope

Electron microscopes can produce more detailed images because of their greater magnification power but also because of their **greater resolution**. The resolution is the smallest distance between two points that can still be seen as two points. So a microscope with a greater resolution can produce a clearer image of objects that are very close together.

CORE PRACTICAL

To use a light microscope, you should:

1. Place the slide on the stage and look through the eyepiece lens
2. Turn the focus wheel to obtain a clear image
3. Start with the lowest objective lens magnification
4. Increase the magnification of the objective lens and refocus.

In order to use specimens with a light microscope, you have to first prepare **the slide**. This is done using the following method:

Take a thin layer of cells from your sample by either **peeling** them off (onion cells) or **using a cotton bud (Animal cells)**

1. Add a small amount of the correct chemical stain (you will be told by your teacher which stain to use). Chemical stains are used to make some parts of the specimen more visible when you look at them through the microscope.
2. Apply the cells to your glass slide by placing them on or wiping the cotton bud against it.
3. Carefully lower a coverslip onto your slide, taking care to avoid air bubbles.

Magnification calculations

Using a microscope to estimate size:

The circular area you can see in a light microscope is called the field of view. If we know its diameter we can then estimate sizes of specimens / parts of specimens.

- Measure your field of view by placing a ruler/graticule on the stage.
- Convert your measurement from mm to μm by multiplying by 1000.
- Count how many cells can be seen across the field of view and divide your field of view measurement by the amount of cells.
- This will give you an estimate of the diameter of one cell.
- Make sure the magnification during both measurements are the same.

You should know how to perform magnification calculations. Remember:

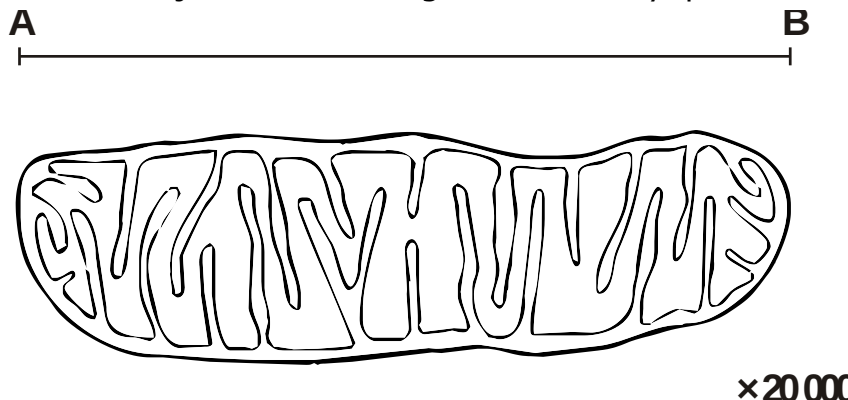
Important calculations

$$\text{Magnification} = \text{Image size} / \text{actual size}$$

$$\text{Actual size} = \text{Image size} / \text{magnification}$$

$$\text{Image size} = \text{Actual size} \times \text{magnification}$$

$$\text{Total magnification} = \text{objective lens magnification} \times \text{eyepiece lens magnification}$$



Calculate the actual length of the organelle above as shown by the line AB in the diagram. Express your answer to the nearest micrometer (μm).

Show your working.

- Measure the line A-B. This is your image size.

$$A = \frac{I}{M} \qquad A = \frac{102\text{mm}}{20\,000} \qquad A = \frac{102000\mu\text{m}}{20\,000}$$

$$\text{Answer} = 5.1. \mu\text{m}$$

Unit Conversions

Due to the microscopic size of specimens we often look at in Biology we tend to use different units than standard, this is to prevent confusion caused by very large numbers below 1.

<u>Unit</u>	<u>Operation</u>	<u>New Unit</u>
Centimetre (cm)	Multiply by 10	Millimetre (mm)
Millimetre (mm)	Multiply by 1000	Micrometre (um)
Micrometre (um)	Multiply by 1000	Nano Metre (nm)
Nano Metre (nm)	Multiply by 1000	Picometre (pm)

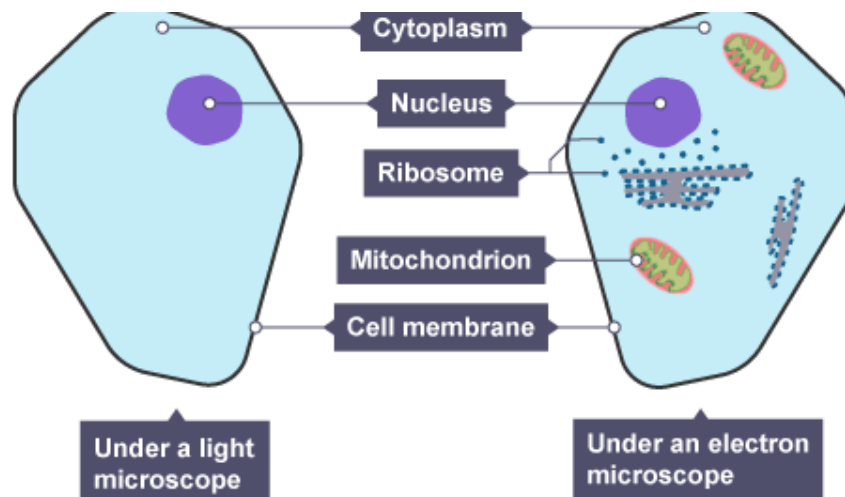
<u>Unit</u>	<u>Operation</u>	<u>New Unit</u>
Nano Metre (nm)	Divide by 1000	Micrometre (um)
Micrometre (um)	Divide by 1000	Millimetre (mm)
Millimetre (mm)	Divide by 10	Centimetre (cm)

Cells

We divide cells into two groups: **Eukaryotic** and **Prokaryotic**

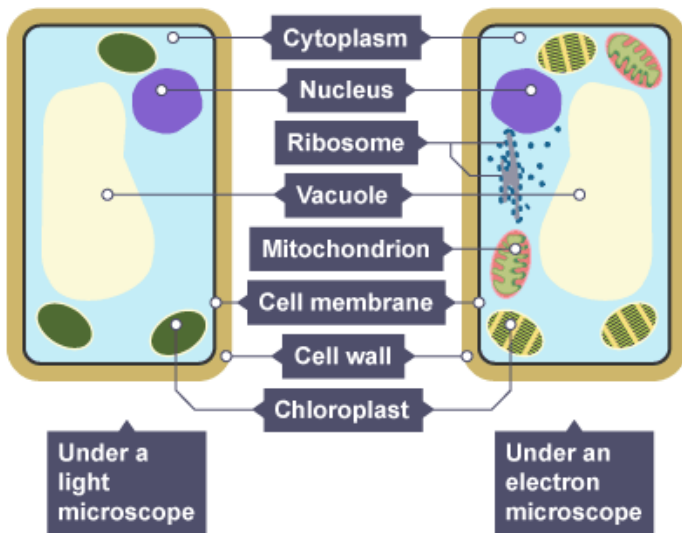
Eukaryotic	Prokaryotic
e.g. animal and plant cells	e.g. Bacteria cells
Membrane bound organelles	No membrane bound organelles
Have a nucleus	No nucleus
Larger than prokaryotic	Smaller than Eukaryotic

Animal cells



Structure	Function
Nucleus	- Contains DNA coding for a particular protein needed to build new cells. - Enclosed in a nuclear membrane.
Cytoplasm	- Liquid substance in which chemical reactions occur. - Contains enzymes (biological catalysts, i.e. proteins that speed up the rate of reaction). - Organelles are found in it
Cell membrane	Controls what enters and leaves the cell
Mitochondria	Where aerobic respiration reactions occur, providing energy for the cell
Ribosomes	- Where protein synthesis occurs. - Found on a structure called the rough endoplasmic reticulum.

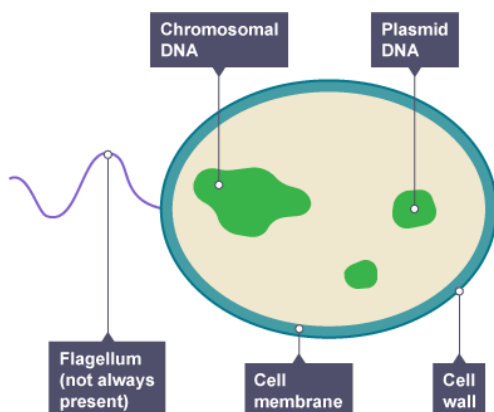
Plant cells



Structure	Function
Chloroplasts	- Where photosynthesis takes place, providing food for the plant - Contains chlorophyll pigment (which makes it green) which harvests the light needed for photosynthesis.
Permanent vacuole	- Contains cell sap - Found within the cytoplasm - Improves cell's rigidity
Cell wall (also present in algal cells)	- Made from cellulose - Provides strength to the cell

Bacterial cells

Bacteria cells are Prokaryotic, so do not have the same features as animal or plant cells



Structure	Function
Cell wall	Made of a different compound (peptidoglycan)
Chromosomal DNA (circular)	As bacterial cells have no nucleus, this floats in the cytoplasm
Plasmids	Small rings of DNA - code for extra genes to those provided by chromosomal DNA
Flagella	Long, thin 'whip-like' tails attached to bacteria that allow them to move

Specialised cells

Examples of specialised cells in animals:

Sperm cells: specialised to carry the male's DNA to the egg cell (ovum) for successful reproduction.

- Streamlined head and long tail to aid swimming.
- Many mitochondria (where respiration happens) which supply the energy to allow the cell to move.
- The acrosome (top of the head) has digestive enzymes which break down the outer layers of membrane of the egg cell
- Haploid nucleus - the word haploid simply means that it has 23 chromosomes, rather than the 46 that most other body cells have.

Egg cells: specialised to accept a single sperm cell and develop into an Embryo

- Surrounded by a special cell membrane which can only accept one sperm cell (during fertilisation) and becomes impermeable following this.
- Lots of mitochondria to provide an energy source for the developing embryo.
- Large size and cytoplasm to allow quick, repeated division as the embryo grows.

Ciliated epithelial cells: specialised to waft bacteria (trapped by mucus) to the stomach

- Long, hair-like processes called cilia waft bacteria trapped by sticky mucus down to the stomach, where they are killed by the stomach acid. This is one of the ways our body protects against illness.

Examples of specialised cells in plants:

Root hair cells: specialised to take up water by osmosis and mineral ions by active transport from the soil as they are found in the tips of roots.

- Have a large surface area due to root hairs, meaning more water can move in.
- The large permanent vacuole affects the speed of movement of water from the soil to the cell.
- Mitochondria to provide energy from respiration for the active transport of mineral ions into the root hair cell.

Xylem cells: specialised to transport water and mineral ions up the plant from the roots to the shoots.

- When they are forming, a chemical called lignin is deposited which causes the cells to die. They become hollow and are joined end-to-end to form a continuous tube allowing water and minerals to move through.
- Lignin is deposited in spirals which helps the cells cope with the pressure from the movement of water

Phloem cells: specialised to carry the products of photosynthesis (sugars/sucrose) to all parts of the plants

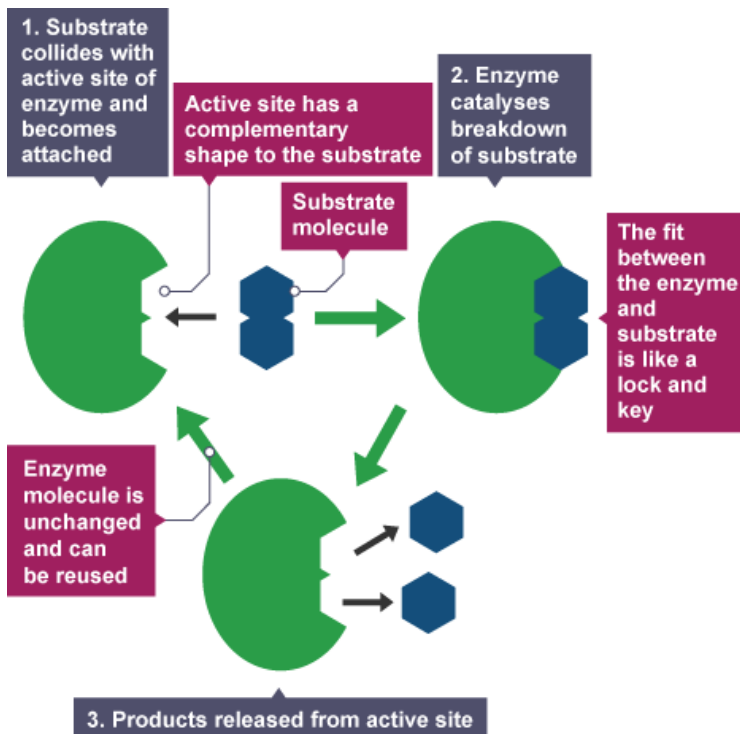
- Cell walls of each cell form structures called sieve plates when they break down, allowing the movement of substances from cell to cell
- Despite losing many sub-cellular structures, the energy these cells need to be alive is supplied by the mitochondria of the companion cells.

Enzymes

Enzymes are proteins that act as Biological Catalysts. This means that they speed up reactions without taking part in or changing as a result of the reaction.

The Lock and Key Hypothesis (a simplified explanation of how enzymes work):

Enzymes are folded into 3d shapes that allow other molecules to fit inside of them. A molecule that fits inside the enzyme is known as the substrate. The substrate fits into the active site of the enzyme.



The shape of the substrate is specific to the shape of the active site on the enzyme (matches the shape of the active site), so when they bond it forms an enzyme-substrate complex.

Once joined, the reaction takes place and the products are released from the surface of the enzyme.

Enzymes can digest (break down substances) or synthesise (join substances together).

Enzymes can only catalyse (speed up) reactions when they bind to a substrate that has a complementary shape, as this is the only way that the substrate will fit into the active site. This is called

enzyme specificity. The diagram shows this happening.

Enzymes in the body

Carbohydrases convert **carbohydrates** into simple **sugars**

Example: amylase breaks down starch into maltose and Glucose. It is produced in your salivary glands, pancreas and small intestine (most of the starch you eat is digested here)

Proteases convert **proteins** into **amino acids**

Example: pepsin, which is produced in the stomach, other forms can be found in pancreas and small intestine.

Lipases convert **lipids (fats)** into **fatty acids** and **glycerol**

Produced in the pancreas and small intestine.

Soluble glucose, amino acids, fatty acids and glycerol pass into the bloodstream to be carried to all the cells around the body. They are used to build new carbohydrates, lipids and proteins, with some glucose being used in respiration. Building these new carbohydrates, lipids and proteins requires energy.

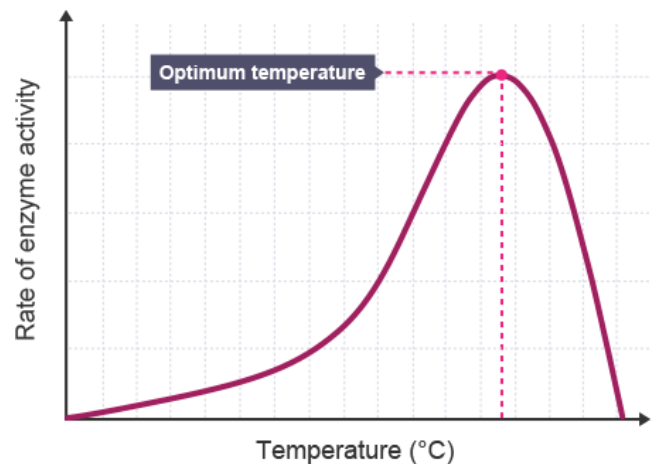
Factors affecting enzyme function

Enzymes are affected by **pH, temperature and substrate concentration**. If enzymes are exposed to extremes of pH or high temperatures the shape of their active site may change. If this happens then the substrate will no longer fit into the enzymes. This means the key will no longer fit the lock. We say that the enzyme has been denatured. It is important you NEVER say that an enzyme has been killed. An enzyme is not alive therefore it cannot be killed it can simply be denatured.

Temperature and enzymes

The rate of reaction increases with an increase in temperature up to an "optimum", but above this temperature it rapidly decreases and eventually the reaction stops.

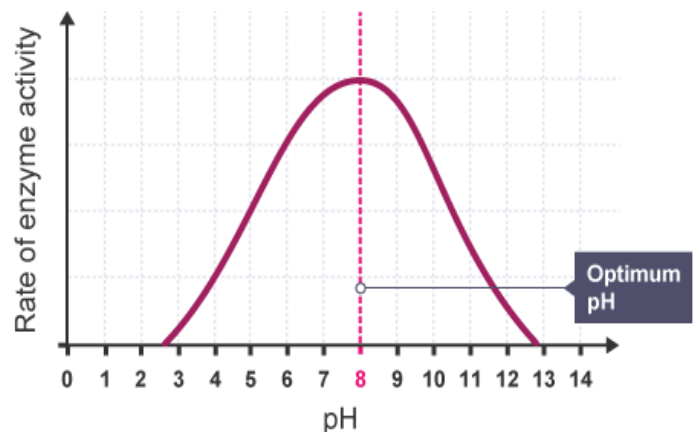
When the temperature becomes too hot, the bonds that hold the enzyme together will begin to break. This changes the shape of the active site, so the substrate can no longer 'fit into' the enzyme. The enzyme is said to be denatured and can no longer work.



pH and Enzymes

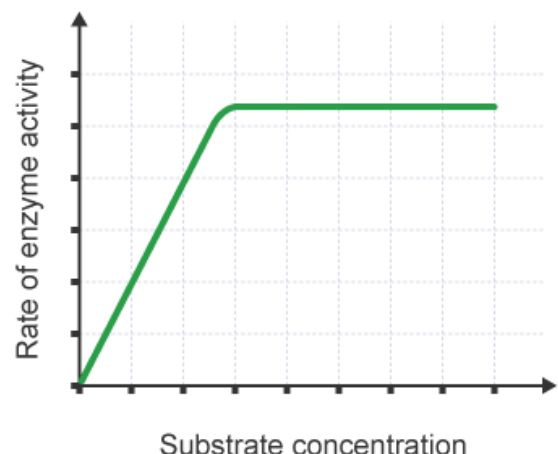
The optimum pH for most enzymes is 7 (neutral), but some that are produced in acidic conditions, such as the stomach, have a lower optimum pH.

If the pH is too high or too low, the forces that hold the amino acid chains that make up the protein will be affected. This will change the shape of the active site, so the substrate can no longer fit in. The enzyme is again said to be denatured, and can no longer work.



Substrate concentration and Enzymes

Enzymes will work best if there is plenty of substrate. As the concentration of the substrate increases, so does the rate of enzyme activity. However, the rate of enzyme activity does not increase forever. This is because a point will be reached when the enzymes become saturated and no more substrates can fit at any one time even though there is plenty of substrate available. This can be seen on the graph where the line showing rate of reaction begins to level off.



CORE PRACTICAL – Investigating pH on enzyme activity

Amylase is an enzyme made in the salivary glands in your mouth and in the pancreas. It catalyses the breakdown of starch into smaller sugar molecules. The iodine test identifies the presence of starch (turns blue/black if starch is present), but does not react with sugar. You will use this test to show how effective amylase is in digesting starch at different pHs.

1. Place one drop of iodine solution into each dimple on the spotting tile.
2. Measure 2cm³ of amylase into a boiling tube
3. Add 2 cm³ of the first pH into the tube
4. Add 2 cm³ of starch solution and place into the water bath.
5. Take a sample every 30 seconds with an empty pipette
6. Add to the iodine, when it stops changing colour stop the experiment.
7. Record the time it took for the colour change to stop happening
8. Repeat using a different pH.

Rate Calculations

Rate calculations are very useful in Biology, and are especially important to determine how fast an enzyme is working (**the rate of reaction**). To perform a rate calculation, we use the formula:

$$\frac{1000}{\text{Time}} \quad \text{OR} \quad \frac{\text{Amount of substrate used/ product formed}}{\text{Time}}$$

Proteases are a type of enzyme used to break down proteins. So as an example, if we added 5g of protein to a solution containing specific protease enzymes, and it took 30 minutes to convert ('use up') all the protein:

Rate = amount of substrate used / time

Rate = 5g / 30 minutes

Rate = 0.17 g/min - this is the rate at which the enzyme is catalysing the reaction, and may change depending on temperature, pH and substrate concentration.

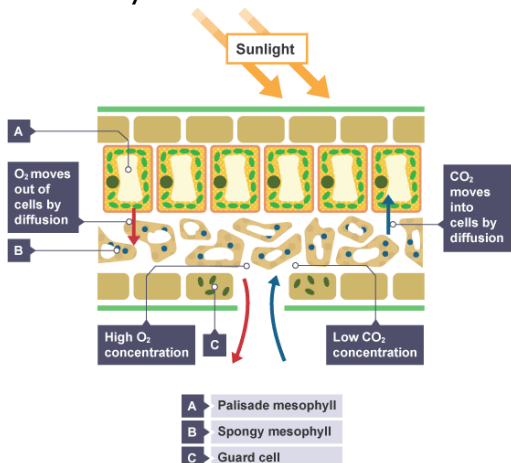
Or

Rate = 5g / 0.5 hours

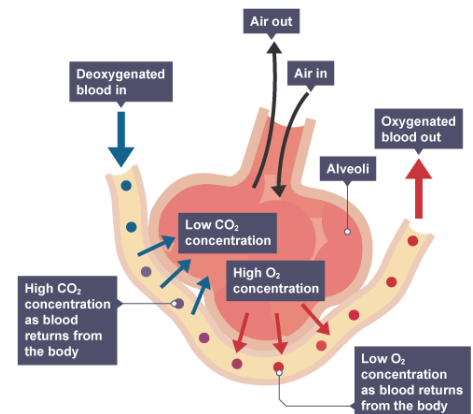
Rate = 10 g/hour - this is the rate at which the enzyme is catalysing the reaction, and may change depending on temperature, pH and substrate concentration.

Transport in cells

Substances like oxygen, glucose and waste products need to be transported in and out of cells constantly to support life processes. This transport generally occurs in one of 3 ways:

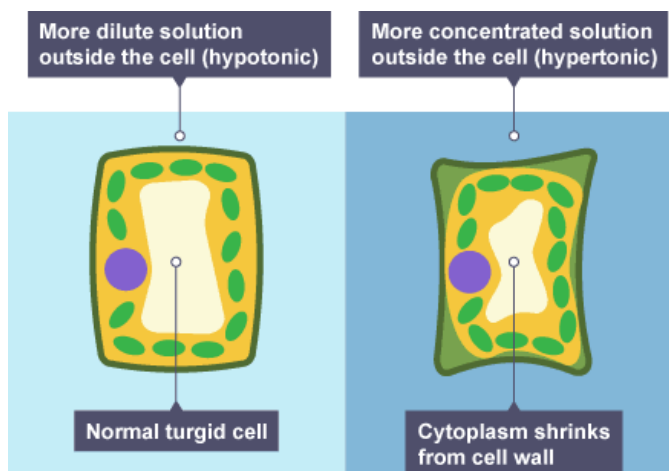


Diffusion - a form of passive transport (does not require energy). It is important to remember that molecules move in every direction and collide with each other, but the net (or resultant) movement is from an area of high concentration to one of low concentration.

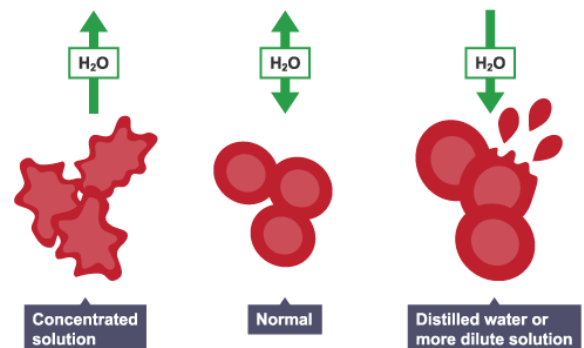


Osmosis is also a form of passive transport (does not require energy) but it only applies to water. The same rules as diffusion apply - however there is no such thing as 'concentration of water', so we say that movement is from a dilute solution to a more concentrated solution, across a selectively permeable membrane. Another way to think about this is that movement of water occurs from an area of low solute concentration to high solute concentration (e.g if the solute was salt, from a less 'salty' solution to a more 'salty' solution).

Osmosis in plant cells

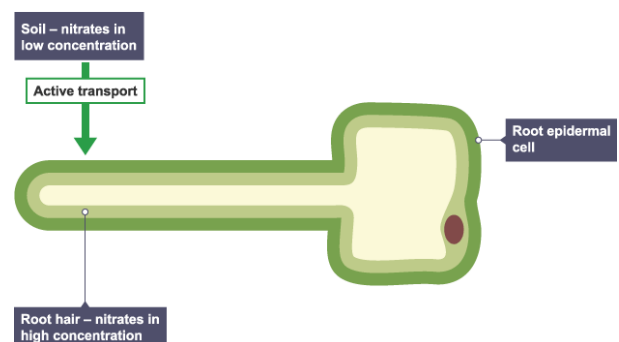


Osmosis in animal cells



Active Transport - active transport is a form of transport that does require energy. This energy comes from ATP,

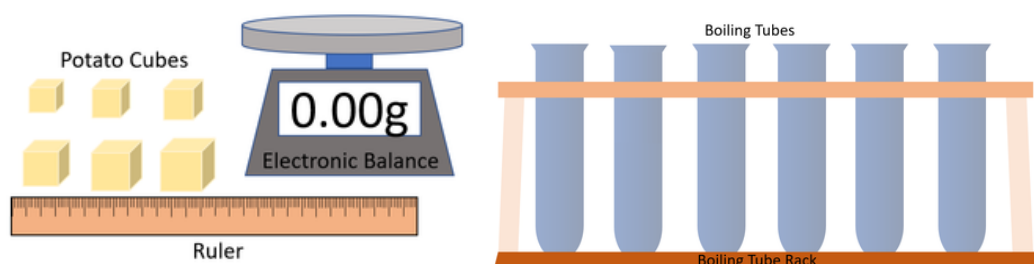
which is the molecule produced in respiration. Active transport is used to move molecules against a concentration gradient (i.e. from an area of low concentration to an area of high concentration). This happens in the root hair cells when the soil has a lower concentration of nitrates than the cell. Active transport is used so that nitrates can continue to be pulled into the cell.



CORE PRACTICAL: Investigating osmosis

1. Prepare sucrose solution; 0%, 20%, 40%, 60% and 80%.
2. Set up a series of boiling tubes with each of these solutions. The 0% sucrose solution will act as the control in the experiment. Make sure each tube is labelled with the concentration.
3. Prepare a blank results table before you begin. Make sure when weighing the potato cylinders, that their masses are not mixed up when recording them. Each cylinder will have a different mass before and after the investigation.
4. Dry a potato strip using a paper towel. Measure the mass of the potato cylinder.
5. Place the potato strip into the 0% solution for 20 minutes.
6. Remove the potato strip, dry it carefully using paper towel. Measure and record the mass of the potato strip.
7. Repeat for each sucrose concentration.
8. For each sucrose concentration, repeat the investigation for several potato cylinders. This allows you to check the precision of your results (results that are close together for the same concentration suggest that the results are precise). The potato cylinders might not all behave in the same way. Making a series of repeat experiments means that any anomalous results can be identified and ignored when a mean is calculated.

This experiment shows the effect of osmosis on plant tissue. The cylinders will decrease or increase in mass if they lose or gain water by osmosis



Percentage change in mass

There is some variation in mass between the potato cylinders at the beginning of the experiment as it would be impractical to prepare the cylinders so that they were identical in mass.

So that we can compare changes in mass of different potato cylinders, it is necessary to calculate the percentage change in mass.

$$\text{change in mass} = \frac{\text{mass at end} - \text{mass at start}}{\text{mass at start}} \times 100$$

Some of the values we obtain for percentage change in mass will be positive, some will be negative, you must include this information in your answers. For the potato cylinder in the distilled water:

$$\text{change in mass} = \frac{2.52 - 2.22}{2.22} \times 100$$

$$\frac{0.30}{2.22} \times 100 = 13.5\%$$

The changes in mass, as percentages, must be calculated for each potato cylinder.

Prove it Questions

Microscopy Pages 2-5

Question	Marks
1. A bacterial cell is a prokaryotic cell, which type of microscope would be best to use to enable you to see a more detailed image?	2
2. Calculate the magnification of a microscope with an objective lens magnification of x10 and an eyepiece lens magnification of x5.	1
3. Why did we discover more about bacterial cells following 1930?	2
4. Compare Electron and Light microscopes.	2
5. Calculate the image size of a specimen if the actual size is 5 μ m and the magnification is x200.	3

Cell Structure Pages 6-7

Question	Marks
1. Describe the difference between eukaryotic and prokaryotic cells.	2
2. Compare and contrast Animal and Bacteria cells	3
3. List the job of the Mitochondria and the Ribosomes	2
4. Compare and contrast Animal and Plant cells	3

Specialised cells Page 8

Question	Marks
1. Define "specialised cell."	1
2. What does Haploid mean?	1
3. List 2 adaptations of the sperm cell.	2
4. Name one specialised plant cell and describe its adaptations	3

Enzymes Page 9

Question	Marks
1. What is the job of the active site on an enzyme?	1
2. An enzyme is a protein, what are the smaller components it is made from?	1
3. What molecule does Amylase break down and what does it break it into?	2
4. Describe the lock and key theory.	3
5. Enzymes are known as biological catalysts – what does a catalyst do?	1

Factors affecting enzyme function Page 10

Question	Marks
1. List three factors that affect enzyme function	3
2. What does denatured mean?	2
3. Describe how temperature affects Enzyme function	3
4. Describe how substrate concentration affects enzyme function	3

Core practical and rates of reaction Page 11

Question	Marks
1. Write the equation to calculate rates of reaction.	1
2. It took 9 minutes for Amylase to break down Starch into Maltose, calculate the rate of reaction.	2
3. Catalase was used to break down a substrate, it produced 30cm ³ of product over 3 minutes. Calculate the rate of reaction.	2
4. List the independent variable and dependent variable you would have when investigating the effect of pH on rate of enzyme activity.	2

Transport in cells and Core practical page 12-13

Question	Marks
1. Diffusion and Osmosis are passive processes, what does this mean?	1
2. What is a key difference between diffusion and Osmosis?	2
3. Why does Active Transport require energy?	2
4. Calculate the percentage change of a potato is its mass prior to being in Sucrose solution was 3g, and its mass following sucrose solution was 4.9g.	2
5. Using your answer to the above question, describe what this means has happened to the potato cells and why?	4

Prove it Mark scheme

Microscopy Pages 2-5

Answer	Marks
1. An electron microscope (1) as it has greater magnification and resolution (1) and a Prokaryotic cell is smaller. (1)	2
2. $10 \times 5 = \times 50$ (1)	1
3. Electron microscopes were invented, (1) allowing us to see greater detail in smaller cells like bacteria. Greater resolution and greater magnification. (1)	2
4. Light microscopes use light rays (1) and have two lenses.(1) Electron microscopes use electrons (1) Electron microscopes have greater magnification powers (1) and greater resolution (1)	2
5. Image size = Actual size x Magnification (1) Image size = 5×200 (1) Image size = 1000 μm (1)	3

Cell Structure Pages 6-7

Answer	Marks
1. Prokaryotic cells are smaller,(1) don't have a nucleus (1) and don't have membrane bound organelles (1) Eukaryotic cells are larger (1), have a nucleus (1) and have membrane bound organelles (1)	2
2. Animal cells have cytoplasm, mitochondria, ribosomes, nucleus, cell membrane, Bacteria cells have a cell wall, (1), no nucleus (1) Plasmid DNA (1) Chromosomal DNA (1) and often have a flagellum (1)	3
3. Mitochondria is the site for respiration (1) Ribosomes are the site for protein synthesis (1)	2
4. Animal cells have cytoplasm, mitochondria, ribosomes, nucleus, cell membrane, plant cells have all of the above(1) but also have chloroplasts (1) Vacuole (1) and a cell wall (1)	3

Specialised cells Page 8

Answer	Marks
1. A cell with an important (special job) that has specific differences to allow it to carry out that job (1)	1
2. Half the amount of chromosomes (23 individual ones instead of 46) (1)	1
3. Tail to help it swim (1) Acrosome on its head to help it penetrate the egg cell (1) streamlined shape to allow it to move efficiently (1) haploid cell (1)	2
4. Root hair cell (1) Large surface area (1) Thin cell walls (1) Xylem cell (1) no top and bottom to the cell (1) tough cell walls to withstand the pressure of liquid moving throughout (1) Phloem cell (1) Sieve plates in cell walls (1)	3

Enzymes Page 9

Answer	Marks
1. It is where the substrate fits into (1) where the reaction happens (1)	1
2. Amino acids (1)	1
3. Starch (1) into Maltose (1) accept Glucose (1)	2
4. Enzyme acts as lock (1) Substrate as key (1) Substrate fits into active site of enzyme due to special shape (1) Enzyme works to either break down or build substrate (1) Due to shape of active site enzymes are specific (only work with one type of substrate (1)	5
5. Speed up reactions without taking part or changing in a reaction (1)	1

Factors affecting enzyme function Page 10

Answer	Marks
1. pH,(1) Temperature (1) and Substrate concentration (1)	3
2. Active site of enzyme has changed shape (1) Meaning the enzyme cannot work with its specific substrate (1)	2
3. Increasing temp initially increases rate (1) until approx. 37.5°C (if enzyme works in the body) this is the optimum rate (1) Following this increasing temp changes the shape of the active site (1), denaturing the enzyme (1) reducing enzyme function (1)	3
4. As the concentration of the substrate increases so does the rate of enzyme activity. (1) However, the rate of enzyme activity does not increase forever.(1) This is because a point will be reached when the enzymes become saturated and no more substrates can fit at any one time even though there is plenty of substrate available. (1)	3

Core practical and rates of reaction Page 11

Answer	Marks
1. Either 1000/ time (1) or amount of product formed /time (1)	1
2. $9 \times 60 = 540\text{s}$ (1) $1000/540 = 1.85\text{s}^{-1}$ (1)	2
3. $3 \times 60 = 180$ (1) $30/180 = 0.17\text{s}^{-1}$ (1)	2
4. IV = pH (1) DV = Time taken for colour change to stop happening (no starch left)	2

Transport in cells page 12

Answer	Marks
1. Neither require energy (1)	1
2. Osmosis refers to the movement of water only, (1) diffusion describes the movement of solute or gases (1)	2
3. Because it moves substances against the concentration gradient (1) From an area of low concentration to an area of high concentration (1)	2
4. $4.9 - 3 = 1.9\text{ g}$ (1) $1.9/3 = 0.63$ $0.63 \times 100 = +63\%$ (1)	2
5. Mass has increased, (1) indicating water has moved from solution into the potato cell (1) Concentration of solution outside the cell was lower (1) (more water outside) than the concentration within the potato cell (less water inside) (1)	4

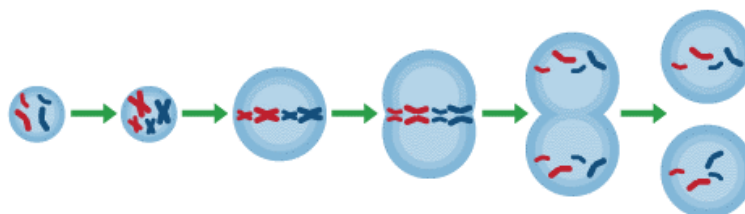
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Topic 2 – Cells and Control

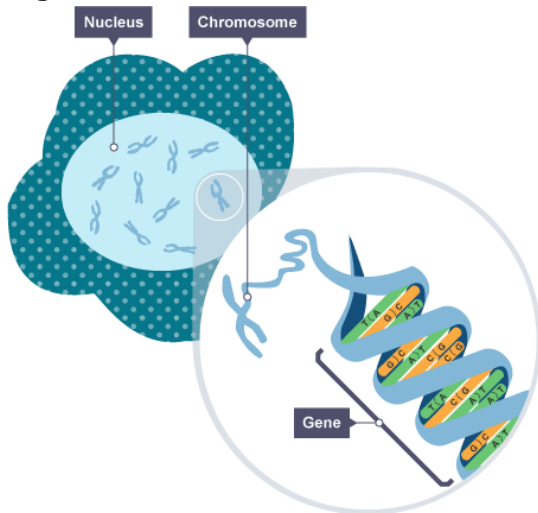
Revision booklet

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Mark scheme	28-29



Cell division

Mitosis is a type of cell division which produces two identical **diploid** daughter cells from one adult diploid cell. For most of the time, DNA exists in the nucleus as thin strands. When the cells containing nuclei are ready to divide, the DNA copies itself then coils and condenses to form chromosomes. Each chromosome is made from a single molecule of DNA. Each section of this single molecule of DNA contains a code for the production of a particular protein called a gene.



Each human body cell contains 46 chromosomes. These can be arranged into 23 pairs. Each chromosome in a pair carries the same types of genes. The 23rd pair is the sex chromosomes. In females, the two chromosomes are identical in shape. These are X chromosomes. Females are referred to as XX. In males, one of the chromosomes is different in shape. This is a Y chromosome. Males are referred to as XY.

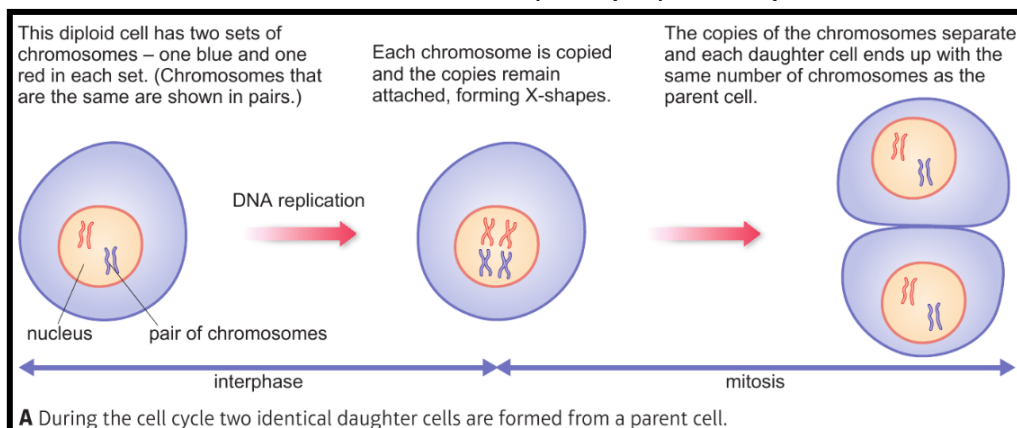
Mitosis

Cells divide when:

- An organism grows.
- An organism becomes damaged and needs to produce new cells for repair.
- Organisms like bacteria reproduce asexually.

It is essential that any new cells produced contain genetic information that is identical to the parent cell.

A growing and dividing cell goes through a series of stages called the cell cycle. Cells spend most of this time cell cycle in 'interphase'. During this phase the cell makes extra organelles, like mitochondria. The chromosomes are also copied (duplicated).



For about 10% of the cell cycle, the cell undergoes a type of cell division called mitosis. In mitosis, two cells called daughter cells are produced, each identical to the parent cell.

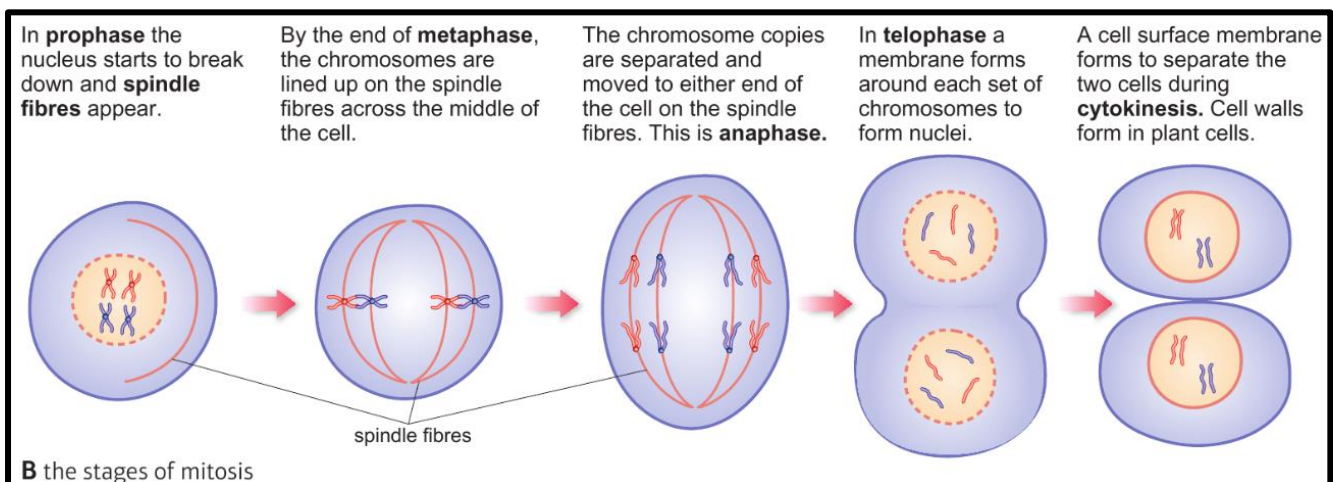
The stages of mitosis in detail

Mitosis is a type of cell division in which a diploid body cell copies itself and finally divides into two identical diploid daughter cells. The daughter cells are clones of each other, every base pair of their DNA is identical.

The stages of mitosis are:

Stage	What happens
Interphase	The cell spends most of its life in this phase. The DNA in chromosomes copies itself ready for mitosis.
Prophase	The DNA in chromosomes and their copies condenses to become more visible. The membrane around the nucleus disappears.
Metaphase	Chromosomes and their copies line up in the middle of the cell.
Anaphase	Chromosomes and their copies are pulled to different ends of the cell.
Telophase	New membranes form around the chromosomes at each end of the cell.
Cytokinesis	The cell membrane pinches in and eventually divides into two daughter cells.

Only two pairs of chromosomes are shown in the diagrams below. There are 23 pairs of chromosomes in a diploid human body cell.



Asexual Reproduction

Asexual reproduction is reproducing young with just one parent. This means the offspring are genetically identical (clones), with identical DNA to the parent.

Asexual reproduction is faster than sexual reproduction but **does not** result in variation as in sexual reproduction.

Examples of organisms that reproduce asexually: strawberry plant (runners); potato plant (tubers)

Cancer

Cells grow then divide by mitosis only when we need new ones. This is when we're growing or need to replace old or damaged cells. The rate at which cells divide by mitosis is controlled by genes in DNA. If a mutation occurs on one of these genes, the cell may start dividing uncontrollably. When a cell becomes cancerous, it begins to grow and divide uncontrollably. New cells are produced even if the body does not need them. A group of cancerous cells produces a growth called a tumour.

Cancer cells are **undifferentiated** – they do not carry out their normal function. At some point, secondary tumours may develop.

Cell differentiation

Animals and plants produced by sexual reproduction begin life as a single cell, a fertilised egg or zygote. These cells divide by mitosis to produce a multicellular organism.

The cells of multicellular animals and plants must also differentiate, differentiation is the process that turns a normal body cell into a specialised cell, specialised cells have developed features that enable them to do specific jobs. (Sperm cell, egg cell, ciliated epithelial cell, red blood cell and many more) Without this specialisation, complex multicellular animals and plants would not exist.

In animals, almost all cells differentiate at an early stage and then lose the ability to differentiate again. Most specialised cells can make more of the same cell by undergoing mitosis (the process that involves a cell dividing to produce 2 identical cells). Others such as red blood cells (which lose their nucleus) cannot divide and are replaced by adult stem cells (which retain their ability to undergo differentiation). In mature animals, cell division mostly only happens to repair or replace damaged cells, as they undergo little growth.

Examples of specialised cells in animals:

Sperm cells: specialised to carry the male's DNA to the egg cell (ovum) for successful reproduction.

- Streamlined head and long tail to aid swimming.
- Many mitochondria which supply the energy to allow the cell to move.
- The top of the head has Acrosome (a digestive enzyme) on it which breaks down the outer layers of membrane of the egg cell, making it easier for the sperm to penetrate the egg.
- Haploid nucleus - the word haploid simply means that it has 23 chromosomes

Egg cells: specialised to accept a single sperm cell and develop into an embryo

- Surrounded by a special cell membrane which can only accept one sperm cell (during fertilisation) and becomes impermeable (does not allow more in) following this.
- Lots of mitochondria to provide an energy source for the developing embryo.
- Large size and cytoplasm to allow quick, repeated division as the embryo grows.

Ciliated epithelial cells: are specialised by having long, hair-like structures called cilia waft bacteria trapped by sticky mucus, to the back of the throat down to the stomach, where they are killed by the stomach acid. This is one of the ways our body protects against illness.

Cells lining small intestine:

- Absorb food molecules produced by digestion. Have tiny folds called micro villi to increase the surface area allowing faster absorption of food.

Some specialised cells of plants:

Cells of the:		Specialised to:
Leaf	Palisade mesophyll	Carry out photosynthesis.
	Spongy mesophyll	Allow gases to circulate for the exchange of gases between the leaf and the environment. Carry out photosynthesis.
	Guard cells	Open and close to control the exchange of gases – carbon dioxide, water vapour and oxygen.
Phloem	Sieve tubes	Transport products of photosynthesis, including sugars and amino acids, from the leaf to where they are needed.
	Companion cells	Provide the energy required for transporting substances in sieve tubes.
Xylem	Xylem vessels	Transport water and dissolved minerals from the roots, up the plant.
Growing points	Meristem	Produce new cells as they divide.

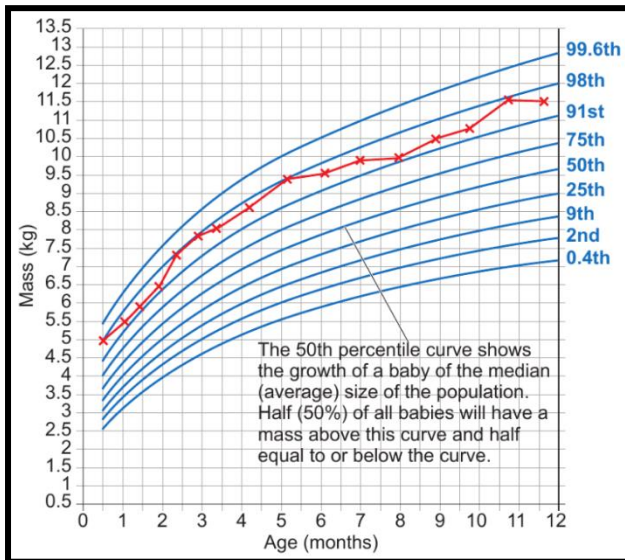
Monitoring growth

The growth of babies is measured in the following ways:

1. Mass (kg)
2. Length (cm)
3. Head circumference (cm)

The growth of babies in all three of these measurements can be monitored using percentile growth charts.

A percentile growth chart for boy's height.



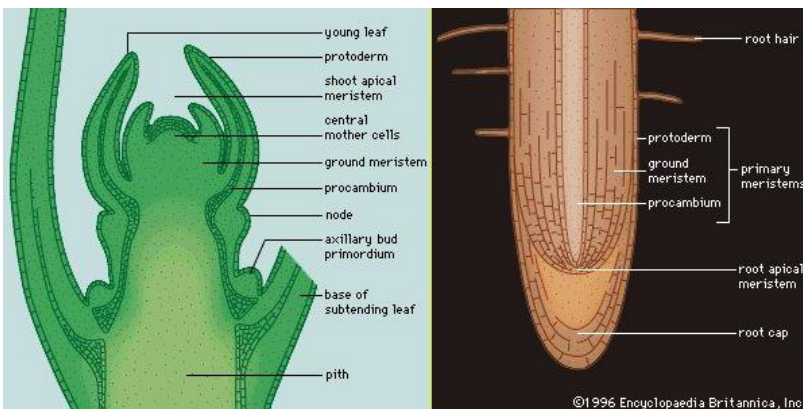
B Percentile growth curves for UK baby boys from 2 weeks to 1 year, for mass. The red line that has been plotted on the curves shows the growth of one baby.

- A baby born at the **50th percentile** for mass is heavier than **50% of babies**.
- A baby born at the **25th percentile** for mass is lighter than **75% of babies**.
- A baby born at the **75th percentile** for mass; 75% of babies in the sample are lighter than or equal to their mass.

Health visitors often visit parents with new babies to take these growth measurements. Health visitors and parents tend to worry more when babies change their position on these charts over time. They would probably be more concerned about a baby that drops from the 50th to the 10th percentile than one that remains on the 10th percentile. This change could be an indication of a health problem.

Plant Growth

Mitosis happens throughout the bodies of animals but only occurs in specific regions of plants. These regions are called meristems and are found in the tips of roots and shoots. Here new cells are formed.



Plant cells are also able to become longer as they grow. This process is called cell elongation and occurs throughout the plant, not just in the meristems. **Cell elongation does not occur in animals.**

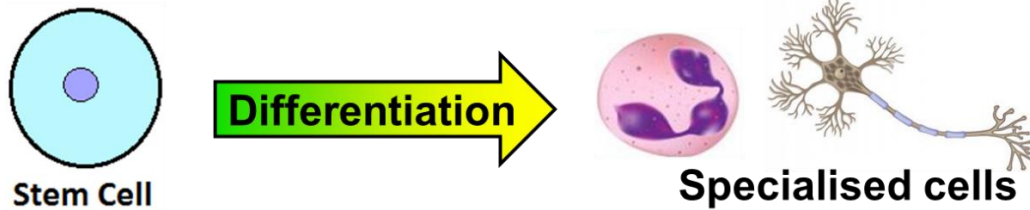
Plant cells can grow longer in a specific direction by absorbing water into their vacuoles, and this is controlled by substances called

auxins. In plants, many types of cells retain the ability to differentiate throughout life. They only differentiate when they reach their final position in the plant, but they can still re-differentiate when it is moved to another position.

Plants have 3 zones of growth; zone of cell division, zone of cell elongation and zone of cell differentiation.

Stem cells

Stem cells are cells that have not undergone differentiation. A cell which has not yet become specialised is called undifferentiated.

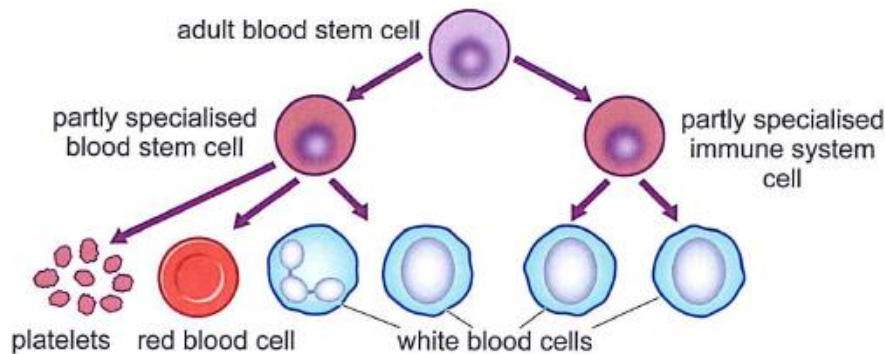


Types of stem cells

1. Embryonic stem cells

- Form when an egg and sperm cell fuse to form a zygote
- They can differentiate into any type of cell in the body
- Scientists can clone these cells (though culturing them) and direct them to differentiate into almost any cell in the body
- These could potentially be used to replace insulin-producing cells in those suffering from diabetes, new neural cells for diseases such as Alzheimer's, or nerve cells for those paralysed with spinal cord injuries

2. Adult stem cells

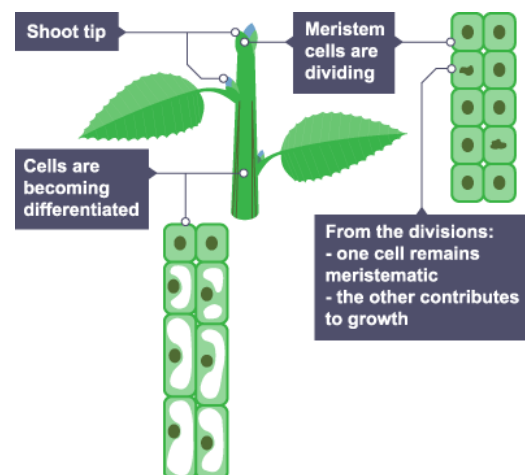


C Blood stem cells are found in marrow in the middle of long bones (such as the femur). They continue to divide throughout life to produce new blood cells.

- If found in bone marrow they can form many types of cells including blood cells. Some adult stem cells remain in the bodies of adults. These are found in limited numbers at certain locations in the body. Adult stem cells can be found in: nose, brain, eyes, blood, heart, liver, bone marrow, skin, muscle
- Adult stem cells can differentiate into related cell types only. So bone marrow cells can differentiate into blood cells and cells of the immune system, but not other cell types.

3. Meristems in plants

- Found in root and shoot tips
- They can differentiate into any type of plant, and have this ability throughout the life of the plant
- They can be used to make clones of the plant- this may be necessary if the parent plant has certain desirable features. Cells of the meristem can differentiate to produce all types of plant cells at any time during the life of the plant. The main meristems are close to the tip of the shoot, and the tip of the root.



Use of Stem cells

Stem cells can divide to produce new cells, which can then divide into different cell types. Stem cells have great potential for:

- Treating patients with currently untreatable conditions, e.g. sickle cell anaemia.
- growing organs for transplants
- medical research

For example:

- in type 1 diabetes
- in cases of multiple sclerosis, which can lead to paralysis
- in cases of spinal cord or brain injury, that have led to paralysis

The stem cells used could be:

- embryonic stem cells
- adult stem cells

Embryonic stem cells can differentiate into a wider range of cell types, but are difficult to obtain. The best source is a five-day-old embryo, although there are clinical, ethical and social issues with their use, as harvesting these cells results in the destruction of the embryo.

Adult stem cells will differentiate into a narrower range of cell types. Bone marrow transplants are an example of adult stem cell transplant. Bone marrow cells will differentiate into different types of blood cell. Bone marrow transplants are carried out:

- in cases of blood cell cancer such as leukaemia and lymphoma
- when blood cells have been destroyed by cancer treatment

Benefits and risks in using stem cells

Benefits	Problems
Can be used to replace damaged or diseased body parts.	We do not completely understand the process of differentiation, so it is hard to control stem cells to form the cells we desire.
Unwanted embryos from fertility clinics could be used as they would otherwise be discarded.	Removal of stem cells results in destruction of the embryo.
Research into the process of differentiation.	People may have religious or ethical objections as it is seen as interference with the natural process of reproduction.
	If the growing stem cells are contaminated with a virus, an infection can be transferred to the individual.
	Money and time could be better spent on other areas of medicine.

Clinical issues

- The difficulty in finding suitable stem cell donors.
- The difficulty in obtaining and storing a patient's embryonic stem cells.
- Cultured stem cells could be contaminated with viruses which would be transferred to a patient.
- Stem cells divide very quickly, if this isn't controlled then a tumour could develop.

- Rejection – if implanted cells aren't grown from patient's cells then the patient's body would recognise them as foreign and their immune system would attempt to destroy the cells.

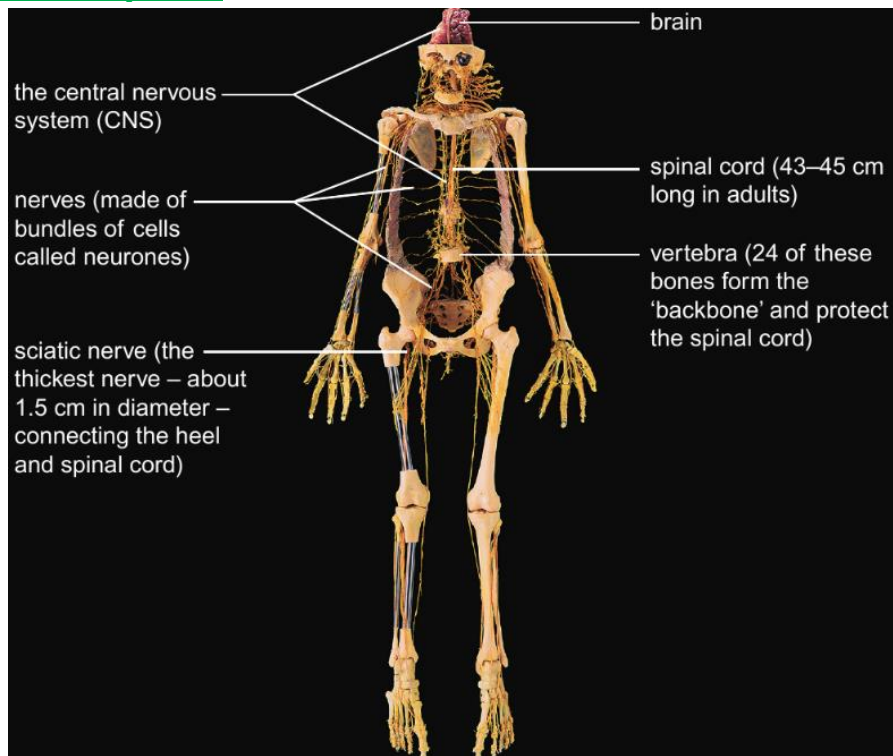
Ethical issues

- A source of embryonic stem cell is unused embryos produced by in vitro fertilisation (IVF)
- For therapeutic cloning, is it right to create embryos just to be used for therapy, and destroy them in the process?
- Embryos could come to be viewed as a commodity, and not as an embryo that could develop into a person.
- At what stage of its development should an embryo be regarded as, and treated as, a person?

The Nervous system

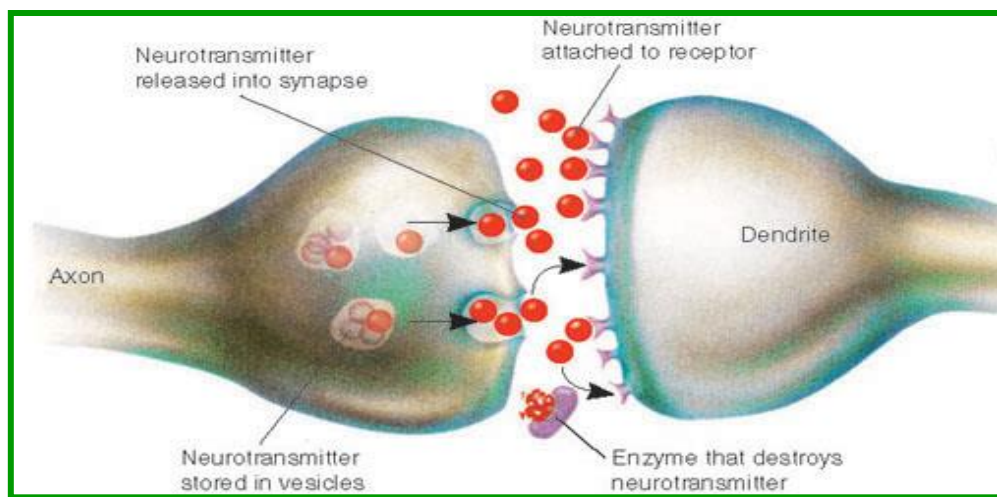
The nervous system controls how you react to what happens around you. It is made up of a system of connecting neurones, receptor cells (found within sense organs) and the brain and spinal cord.

The Central Nervous System



Receptor cells detect a change in stimuli in your environment (eg temperature.) Different receptor cells detect different stimuli – e.g. eyes detect light, nose detects smell etc. This information is changed to an electrical impulse which is sent along sensory neurones to the brain. The brain coordinates an appropriate response, this information is then sent along relay neurones (in the CNS) to motor neurones to an effector (muscle or gland). The effector then acts accordingly.

Synapses



Synapses are the gaps between neurones. When an impulse reaches a synapse the axon terminal releases a chemical called a neurotransmitter which is diffused across the gap to receptors found on the dendrites of the next neurone. This sets off a new electrical signal in the next neurone. This process slows a message down as the diffusion of neurotransmitters takes a long time.

Neurones

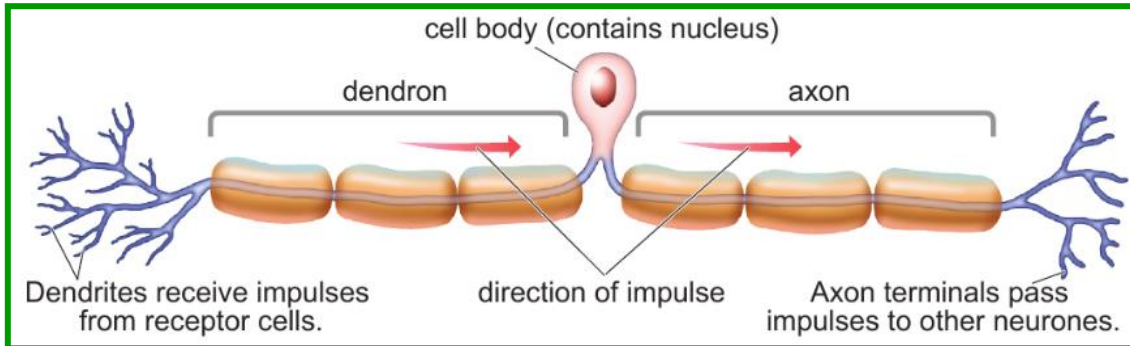
3 types of neurones - All neurones have a cell body with a nucleus.

They have extensions to connect to other neurones – dendrites, etc.

They can be very long which speeds up the message, (connecting with another neurone slows a message down) so the longer the neurone the less connections they will need.

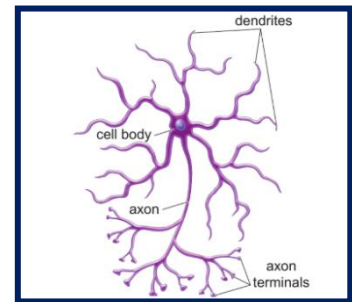
Sensory neurones

One long Dendron carrying electrical impulses from receptor cells to cell body (which is in the middle of the neurone) then along the axon to the CNS. Has myelin sheath surrounding the axon and Dendron to insulate the impulse travelling through the middle – this prevents interference and allows the message to continue moving quickly.



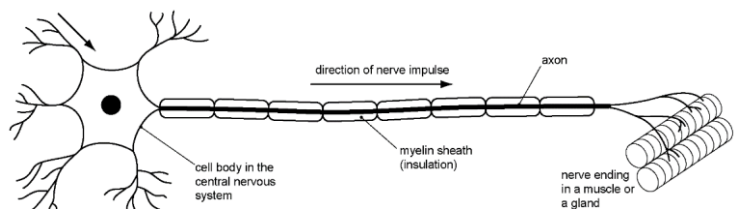
Relay Neurone

Short dendrites carry impulses from sensory neurones to the cell body. Axon carries nerve impulses to the motor neurones. Does not have a Dendron.

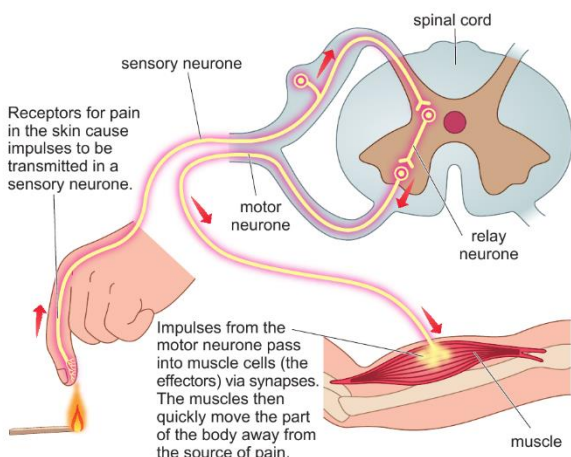


Motor Neurone

Short dendrites carry nerve impulses from CNS to the cell body. Then the long axon carries nerve impulses to effector cells. No Dendron. Has myelin sheath surrounding the axon and Dendron to insulate the impulse travelling through the middle – this prevents interference and allows the message to continue moving quickly.



Reflex arc



Not all movements are linked to the CNS, reflexes are automatic responses to stimuli that can reduce the chance of injury.

Stimuli is detected by the receptor cells, impulse is passed down a sensory neurone, directly to a relay neurone in the spinal cord. The impulse then continues to a motor neurone and then to an effector. Because a reflex arc does not involve the brain and you do not have to spend time thinking about it, it is quicker than a normal response.

Prove it Questions

Page 19-20 Mitosis	
1. What is Mitosis and what does it produce?	(2)
2. List 3 uses of Mitosis in organisms.	(3)
2. Name and describe the stages of Mitosis	(6)
3. Explain how Mitosis can be linked to uncontrollable cell division.	(3)

Page 21 Cell differentiation and specialised cells	
1. What does cell differentiation mean?	(2)
2. What types of cell can differentiate, and how do they differ?	(4)
3. Give examples of 2 specialised plant cells and explain how they are adapted to do their job.	(4)
4. Give examples of 2 specialised animal cells and explain how they are adapted to do their job.	(4)

Page 22 Measuring Growth	
1. How do animals grow?	(2)
2. Referring to the chart on page 7, describe the growth pattern of a boy on the 5 th percentile line between ages 2- 20.	(3)
3. A child at age 2 is on the 50 th percentile line, what does this mean?	(2)
4. A health visitor notices that a child has dropped from the 50 th percentile to the 10 th percentile in 1 year – would this cause them concern and why?	(4)

Page 23 Stem cells	
1. Name and describe the 2 types of Stem cell found within animals.	(3)
2. Describe how meristems in plants are like embryonic stem cells in animals.	(3)
3. Describe what happens in the each zone of growth in the plant.	(3)
4. Compare plant growth with animal growth.	(3)

Page 24 Use of Stem Cells	
1. List 3 uses of Stem Cells in medicine.	(4)
2. Describe 3 risks of using Stem cells.	(3)
3. Describe 3 benefits of using Stem cells.	(3)
4. Describe 3 ethical issues with the use of embryonic stem cells.	(4)

Page 25-26 The Nervous system and Neurones	
1. List the organs involved in the central nervous system (CNS)	(4)
2. Describe each stage involved in coordinating a response to change in temperature (CNS response).	(4)
3. Describe each stage of how the bodies' nervous system responds to a bee sting.	(4)
4. Compare and contrast the sensory neurone and the motor neurone.	(4)

Mark scheme

Page 19-20 Mitosis	
1. Mitosis is cell division (1) Producing 2 diploid daughter cells (1) from 1 adult cell.	(2)
2. Growth (1). Repair (1) Asexual reproduction (1)	(3)
2. Interphase – DNA replication (1) Prophase- Chromosomes become shorter and more visible (1), membrane surrounding nucleus breaks down (1). Metaphase Chromosomes line up in centre of cell (1) Anaphase – Spindle fibres pull cell apart, (1) they go to opposite sides of the cell (1) Telophase – Membranes form around the new sets of chromosomes (1) Cytokinesis -Cytoplasm and cell membrane divide to become 2 new sets of cells (1)	(6)
3. If a mutation occurs (1) a cell may begin to divide uncontrollably (1) this can cause a mass of abnormal cells (1) called a tumour (1) The tumour can invade and destroy living tissue (1) this is called cancer (1)	(3)

Page 21 Cell differentiation and specialised cells	
1. Differentiation is the process by which cells become specialised, (1) and develop specialist functions.	(1)
2. Stem cells can differentiate (1) Adult stem cells can differentiate into many but not all types of cell (1) Embryonic stem cells can differentiate into all types of cell (1)	(3)
3. Any from table of plant cells listed. Must have job and one adaptation each.	(4)
4. Any from the animal cells listed. Must have job and one adaptation each.	(4)

Page 22 Measuring Growth	
1. Cell division – Mitosis (1)	(1)
2. At age 2 the child is approximately 90cm tall. (1) Growth continues at a consistent rate until age 8,(1) where growth slows slightly until age 12.(1) At this age the rate of growth increases until approx. age 18(1) where it slows very quickly and stops at age 20. (1) The height at this age is approximately 170 cm. (1)	(3)
3. Either 50 % of the sample are below (1) or the same height (1) as them. OR 50% of the sample are above (1) or the same height (1) as them.	(2)
4. This is an inconsistent rate of growth (1) A child should continue growing at a rate consistent with their percentile line (1) give or take one line (1). Dropping from 50 th to 10 th indicates there may be an underlying health problem which would need to be investigated. (1)	(4)

Page 23 Stem cells	
1. Embryonic stem cell (1) Allows cells to differentiate into any type of cell (1) Adult Stem cell (1) allows cell division to many but not all types of cells (1).	(3)
2. Meristems in plants produce unspecialised cells that can differentiate into any type of plant cell. (1) This is like Embryonic stem cells as they can also differentiate into any type of cell. (1) Unlike human stem cells – meristems can continue to divide and differentiate into any type of cell for as long as the plant lives. (1)	(3)
3. Zone of cell division – Mitosis. (1) Zone of cell elongation – Cells expand and get longer (1) increasing the height of the plant. (1) Zone of cell differentiation – Cells become specialised. (1)	(3)
4. Animals grow by Mitosis (1) – simply gaining more cells through cell division. (1) Plants growth through mitosis (1), cell elongation (1) and differentiation. (1) Animals stop growing in adult hood. (1) Plants can continue growing as long as they live. (1) Plants can regenerate new parts. (1) Animals cannot. (1) ANSWER MUST REFER TO MITOSIS.	(5)

Page 24 Use of Stem Cells	
1. Growing organs for transplants (1) Research (1) Treating incurable conditions (1) ANY OTHER JUSTIFIABLE MEDICAL CONDITION.	(3)
2. Any 3 reasons from list of relevant pages. Must include description not simply a list. 1 mark per reason described.	(3)
3. Any 3 reasons from list of relevant pages. Must include description not simply a list. 1 mark per reason described.	(3)
Any 3 reasons from list of relevant pages. Must include description not simply a list. 1 mark per reason described.	(4)

Page 25-26 The Nervous system and Neurones	
1. Sense organs (1) Receptor cells (1) Neurones (1) Brain (1) Spinal cord (1) Effector (1) Accept named sense organs for one mark only. Accept named neurones. Accept muscles of glands for effectors	(4)
2. Receptor cells detect a change in stimuli, (1) impulse transmitted through sensory neurone, (1) to the CNS, (1) through a relay neurone, (1) through a motor neurone, (1) to an effector which carries out the response. (1)	(5)
3. Reflex arc (1) Receptor cells detect a change in stimuli, (1) impulse transmitted through sensory neurone, (1) through a relay neurone, (1) through a motor neurone, (1) to an effector which carries out the response. (1) No brain involvement (1)	(5)
4. Sensory neurone has a Dendron – the Motor neurone doesn't. (1) Sensory neurone has a cell body in the middle of the neurone. Motor neurone has a cell body at the end. (1) Both have myelin sheath surrounding the axon (1) Both have dendrites (1) Both are long (1)	(4)
5. When the impulse reaches a Synapse which is a gap between neurones (1) a neurotransmitter is released (1) this travels across the gap (1) and is picked up by receptors on the next neurone (1) This begins a new electrical impulse (1) This chemical transmission slows the message down slightly. (1)	(4)

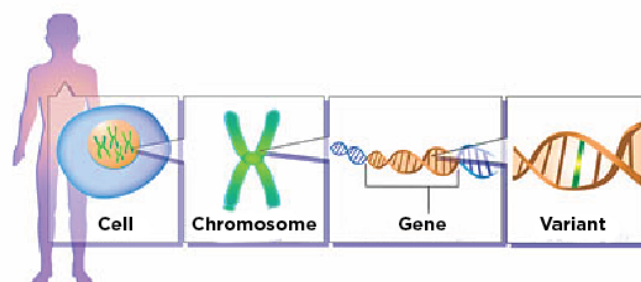
GCSE 9-1 BIOLOGY



Topic 3 – Genetics

Revision booklet

Topic	Pages
Reproduction and Meiosis and DNA	31
DNA extraction and inheritance	32
Punnet Squares	33
Calculations and pedigree analysis	34
Variation	35
Mutation	36
The Human Genome Project	37
Questions	38-39
Mark scheme	40-41



Credit: Adapted from National Library of Medicine

Reproduction and Meiosis

Sexual reproduction

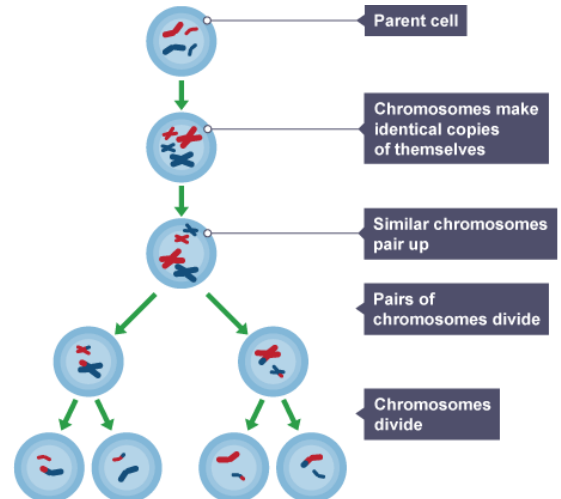
Sexual reproduction involves the joining of two sex cells, or gametes during fertilisation. Organisms produced by sexual reproduction have two parents and are genetically similar to both but not identical to either.

Asexual reproduction only involves one parent so there is no joining of sex cells during fertilisation. Organisms produced by asexual reproduction are genetically identical to each other and their parent. They are clones.

Meiosis (a type of cell division)

There are two types of cell division called mitosis and meiosis. Mitosis produces identical diploid body cells for growth and repair (covered in topic 2). Meiosis produces **haploid** non-identical sex cells, or gametes. These fuse to form a **diploid** fertilised egg cell during fertilisation. Meiosis is different to mitosis as meiosis has two cell divisions, whereas mitosis only has one cell division.

Meiosis produces sperm and egg cells in animals, and pollen and egg cells in plants.



DNA

The genetic material in the nucleus of a cell is made up of a chemical called DNA. (**Deoxyribonucleic acid**). DNA is a **polymer**, made of many smaller units called **nucleotides**.

A **nucleotide** is made of a sugar and a phosphate group, with one of four different **bases, A, C, T or G**, attached. The **nucleotides** join together, forming two strands. These, in turn, form a **double helix** structure. **Weak hydrogen bonds** hold the **complementary base pairs together**. Base **A always pairs with T, and C always pairs with G** forming a twisted ladder structure called a double helix. It carries the genetic code, which determines the characteristics of a living organism.

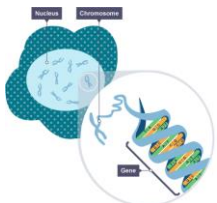
Chromosomes

The cell's nucleus contains chromosomes. These are long threads of DNA, which are made up of many genes. Each cell in your body contains 46 chromosomes (23 pairs) except for the gametes which only contain 23 individual chromosomes. During fertilisation the two gametes will join together to produce cells which have the full amount of chromosomes. This allows for genetic variation to occur. A male has the alleles XY and a female has the alleles XX.

If a Karyogram (a picture of chromosomes) has XY then it is a male. If it contains XX then it is a female. Remember – all cells have 46 chromosomes except for the sex cells (gametes – sperm and egg.)

Genes

A gene is a small section of DNA in a chromosome. Each gene codes for a particular sequence of amino acids in order to make a specific protein. The diagram shows the relationship between the cell, its nucleus, chromosomes in the nucleus, and genes.



Genomes

The genome is one copy of all an organism's DNA. In humans, this is all the DNA that makes up the 23 pairs of chromosomes found in all diploid body cells. That is all the cells except sex cells or gametes, which only have half of a person's genome.

Extracting DNA from fruit

It is possible to extract DNA from cells in a variety of ways. One of the simplest methods is to extract it from fruit like a kiwi.

Aim

The aim of this experiment is to extract DNA from a kiwi fruit.

Method

1. Peel the skin from half a kiwi fruit and mash it up (break down the cell walls)
2. Mix a teaspoon of salt and small volume of washing up liquid into the fruit (release the DNA from the nuclear membrane)
3. Gently heat this mixture at about 60°C for five minutes
4. Filter the mixture and retain only the filtrate (the filtered liquid) (Removes insoluble material)
5. Cool using an ice bath and gently pour chilled ethanol onto the top of the filtrate. (Precipitate the DNA from the solution.)

You will see strands of white DNA with bubbles in them at the boundary between the filtrate and the chilled ethanol.



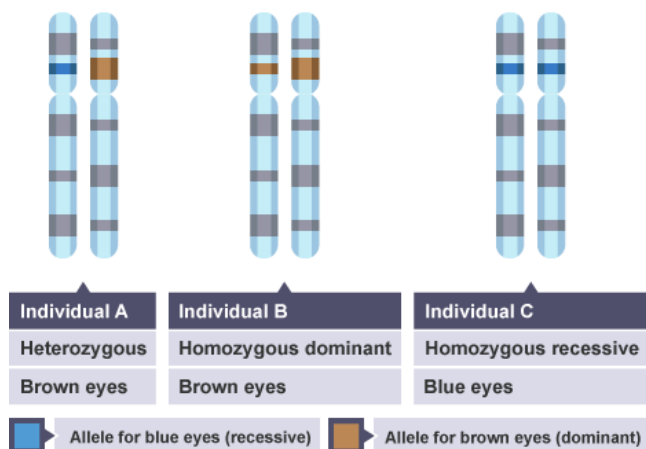
Inheritance

Most characteristics are a result of multiple genes interacting, rather than a single gene.

- Alleles are different versions of the same gene. For example, the gene for eye colour has an allele for blue eye colour and an allele for brown eye colour. For any gene, a person may have the same two alleles, known as **homozygous** or two different ones, known as **heterozygous**.
- The genotype is the collection of alleles that determine characteristics and can be expressed as a **phenotype**. You can see someone's phenotype – e.g. blue eyes, but you can't see their genotype (this is the instruction that produces the characteristic known as the phenotype.)
- Males have the alleles XY
- Females have the alleles XX

The male and female alleles (XY and XX) should be used in a punnett square when being asked to determine probability of sex.

Alleles may be either **dominant** or **recessive**:



- A dominant allele is always shown in the phenotype, even if one copy is present. Dominant alleles are represented by a capital letter, for example you could use a B. The allele for brown eyes, B, is dominant. You only need one copy of this allele to have brown eyes. Two copies will still give you brown eyes.

- A recessive allele is only expressed if the individual has two copies and does not have the dominant allele of that gene. Recessive alleles are represented by a lower case letter, for example, b. The allele for blue eyes, b, is recessive. You need

two copies of this allele to have blue eyes, e.g. bb.

- Homozygous alleles are both identical for the same characteristic, for example BB or bb.
- Heterozygous alleles are both different for the same characteristic, for example Bb.

Genetic cross diagrams (Punnett squares)

Monohybrid crosses

Genetic crosses of single gene combinations (monohybrid inheritance) can be shown and examined using Punnett squares. These show the possible offspring combinations that could be produced, and the probability of these combinations can be calculated.

Example of a genetic cross 1

The height of pea plants is controlled by a single gene, which has two alleles: tall and short.

- The tall allele is dominant and is shown as T.
- The small allele is recessive and is shown as t.

The Punnett square shows the possible allele combinations of the offspring produced when two pea plants are bred.

Parent 1 is TT - tall

Parent 2 is tt - short

Note: You should always write the dominant allele first. This means that the phenotype of all offspring produced will be tall. (100% chance of being tall)

	T	T
t	Tt	Tt
t	Tt	Tt

Example 2

In this genetic cross, Parent 1 is still tall but is heterozygous with alleles Tt. Parent 2 is short and is homozygous recessive with alleles tt.

In this Punnett square the top row shows the female alleles and the left-hand column shows the male alleles.

Half of the possible offspring have the allele combination Tt and will be tall and the other half have the combination tt and will be short.

	T	t
t	Tt	tt
t	Tt	tt

These examples are single gene combinations, but remember that most phenotypes are controlled by multiple genes.

How to construct Punnett squares

1. Find out the parental genotypes. You can use any letter you like (except for when determining sex of offspring) but select one that has a clearly different lower case, for example: Aa, Bb, Dd.
2. Split the alleles for each parent and add them into your Punnett square around the edges.
3. Work out the new possible genetic combinations inside the Punnett square.

You may be asked to comment on the proportion of different allele combinations in the offspring, calculate a probability, ratio or just determine the phenotypes of the offspring.

Example

The inheritance of fur colour in mice is controlled by a single gene. White fur is recessive. Grey fur is dominant. Two mice that are heterozygous for the fur colour gene mate.

Determine the probability that the offspring will have grey fur.

Parent 1 - Aa

Parent 2 - Aa

In this Punnett square the top row shows the alleles of parent 1 and the left-hand column shows the alleles of parent 2.

The genotypes AA and Aa both result in the grey fur phenotype.

The genotype aa results in the phenotype white. The grey and the white are known as 'offspring phenotypes'.

There is a **75% probability** that the offspring will have grey fur.

We also write this as a ratio:

The ratio of grey to white fur is 3:1

	A	a
A	AA grey	Aa grey
a	Aa grey	aa white

Scientific calculations

Using probability, direct proportion and simple ratios

You can express the outcome of a genetic cross using probability (percentages), direct proportion or ratios. It is important to remember during the process of fertilisation, the allele combinations created are a random process, and that is why probability is used, as nothing is guaranteed. **Each of the four possible offspring combinations is as likely to happen during every fertilisation event.**

So, a couple having 4 children does not mean the each child will represent one of each phenotypes listed.

In this Punnett square the top row shows the female alleles and the left-hand column shows the male alleles.

	a	a
A	Aa	Aa
a	aa	aa

Direct proportion

Half the offspring have the combination Aa and half have aa.

OR

Out of 4 offspring 2 have the combination Aa and 2 have aa.

Probability

The probability of the offspring being Aa is 50%.

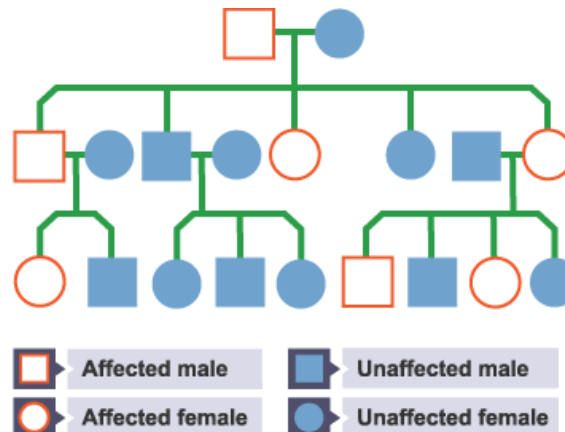
The probability of the offspring being aa is 50%.

Ratio

The ratio of Aa to aa is 1:1.

Pedigree analysis

Doctors can use a pedigree analysis chart to show how genetic disorders are inherited in a family. They can use this to work out the probability that someone in a family will inherit a condition. A pedigree analysis is usually undertaken if families are referred to a genetic counsellor following the birth of an affected child, or if there is a known genetic disorder in the family.



The pedigree analysis chart is used to show the relationship within an extended family. Males are indicated by the square shape and females are represented by circles. Affected individuals are red and unaffected are blue. Horizontal lines between males and females show that they have produced children.

This analysis shows both male and female are affected, and every generation has affected individuals. There is one family group that has no affected parents or children, but the remaining two families have one affected parent and affected children too.

Variation

Variation is defined as the differences seen between individuals of the same species.

Extensive genetic variation is visible within any species. Variation within genes leads to different genotypes, and this can be seen in a different phenotype. Genetic variation and environmental variation can both cause these different phenotypes.

Genetic variation is caused by reproduction and mutation. Examples of genetic variation include blood type, genetic diseases (sickle cell, cystic fibrosis amongst others), eye colour, and natural hair colour.

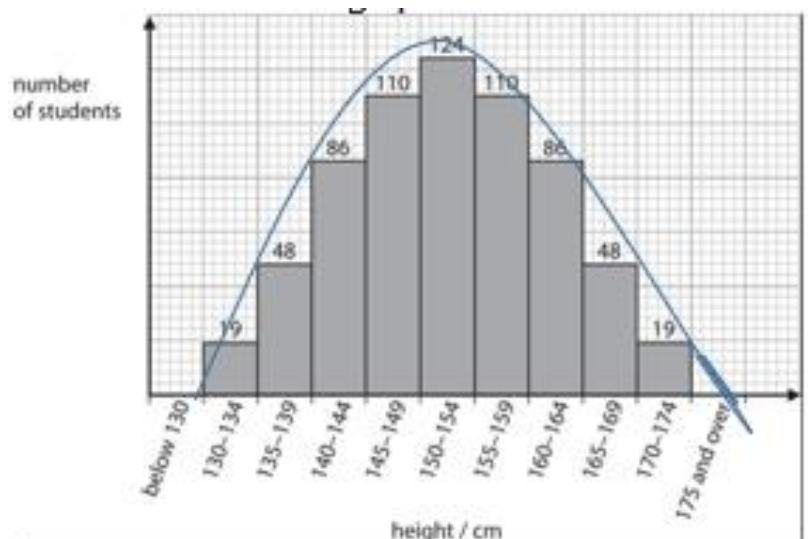
Environmental variation is caused by changes in an environment. Examples of environmental variation include accent, scars, and tattoos.

Some variation is caused by **both the environment and genetics** such as intelligence, height, weight.

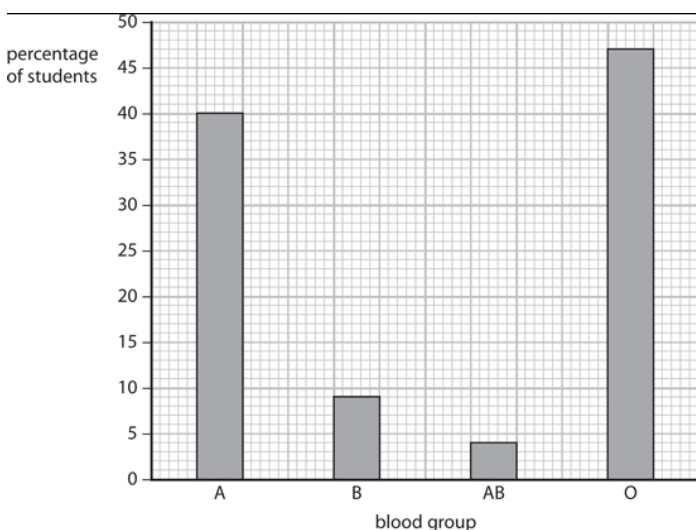
Data summarising variation can be **continuous** or **discontinuous**. Discontinuous data is qualitative and categorical and has distinct divisions between the group, such as blood type or eye colour. Continuous data is generally quantitative (numerical) and can be anywhere within a range of values (e.g. height)

Continuous and discontinuous data

Continuous data will be displayed as a histogram, with frequency groupings. The bars should have no gaps between them – the area of the bar is important here. See below. The histogram here → also shows a normal distribution of data – this is where the frequency at the lower and upper end of the scales is lower than the frequency seen in the middle scales. If you draw a line over the top of the bars, it will create a bell shape. What this means in terms of the example above (height) is that the majority of the people are average height; this is noticed frequently within data collection.

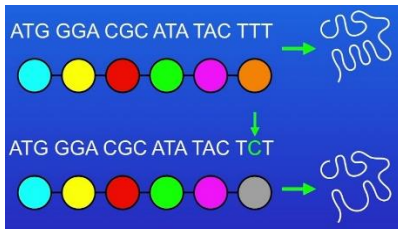


Discontinuous data will be displayed as a bar chart, with spaces between the bars.



Causes of mutation

Mutation occurs continuously and can be spontaneous. It can also happen because of exposure to ionising radiation or exposure to chemical mutagens, such as tar from cigarette smoke.



A mutation is a change in a gene or chromosome: addition (adding an extra base in), deletion (deleting a base) or substitution (swapping a base).

The change in a base sequence in DNA can lead to a change in a protein, which is either faulty or different to what is expected.

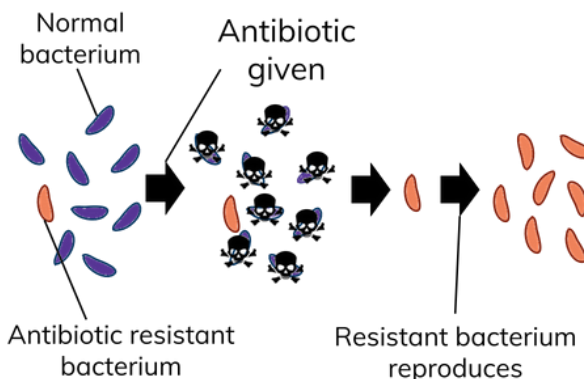
The impact of this could mean that an important enzyme is not made; this would result in a specific substrate not being able to fit into the substrate-binding site. If it is a structural protein such as collagen, it might lose its strength.

However, most DNA mutations do not significantly change a protein; they only alter it slightly, or not at all, so its appearance or function is not changed.

Occasionally, mutations can be positive, give an organism an advantage, or negative, and give a disadvantage but mutations that have a significant effect are rare. Some mutations may have a small effect, but most mutations have no effect on the organism.

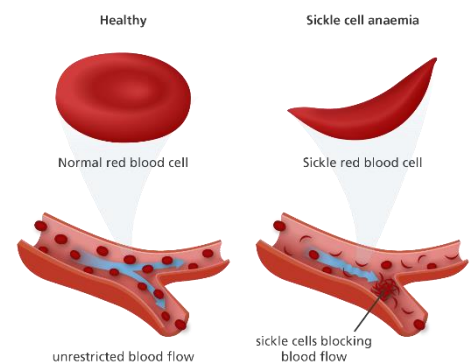
Mutation can cause antibiotic resistance in bacteria

Bacterial strains can develop resistance to antibiotics. This happens because of natural selection. In a large population of bacteria, there may be some cells that are not affected by the antibiotic. These cells survive and reproduce, producing even more bacteria that are not affected by the antibiotic.



Sickle cell anaemia is a blood disease in which red blood cells reveal an abnormal crescent (or sickle) shape when observed under a microscope. It is a recessive inherited disorder.

Only those individuals that inherit two copies of the sickle mutation (one from their mother and the other from their father) develop sickle cell anaemia. If untreated, these individuals have a shorter than normal life expectancy and as such it would be **expected** that this mutation would be rare in human populations. This is however, far from being the case. Observations made during the mid-20th century revealed that the sickle mutation is, in fact, highly, selected in populations from areas of the world where malaria is very frequent, with sometimes 10-40% of the population carrying this mutation. The sickle cell haemoglobin gives a survival advantage against malaria.



The Human Genome Project

The Human Genome Project, or HGP for short, was started at the end of the last century. It had several aims, including:

- to work out the order or sequence of all the three billion base pairs in the human genome
- to identify all the genes
- to develop faster methods for sequencing DNA

The genome of an organism is the entire genetic material of that organism. Each of your diploid body cells (not sperm, eggs or red blood cells) has one copy of your entire genome.

The Human Genome Project started in 1990 and was completed in 2003. Hundreds of scientists from different universities all over the world collaborated to determine the sequence of base pairs that made up the genome of a random man and woman.

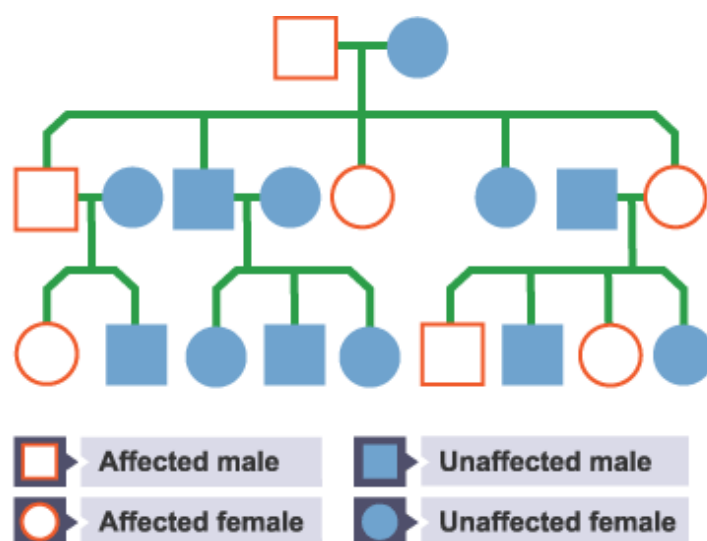
There are more than three billion of these. The mapping of the whole human genome has great importance for medicine. It enables us to:

- search for genes linked to different types of disease
- understand inherited disorders and their treatment
- trace human migration patterns from the past

Scientists are now searching for disease associated genes within the human genome. Two examples of these are genes that can contribute to breast cancer, which are known as *BRCA1* and *BRCA2*. Mutations in these genes account for approximately 10% of all inherited breast cancer cases detected.

Scientists detected *BRCA1* and *BRCA2* genes by studying families where breast cancer was known to have been inherited between individuals. They were able to create a pedigree analysis, which is similar to a family tree diagram that showed the close relationship of those affected and unaffected within the family.

The pedigree analysis illustrates the inheritance pattern of the disease to be determined. This enabled scientists to test DNA from the affected and unaffected individuals to identify differences. It is now possible to detect the presence of the genes by having a simple blood test.



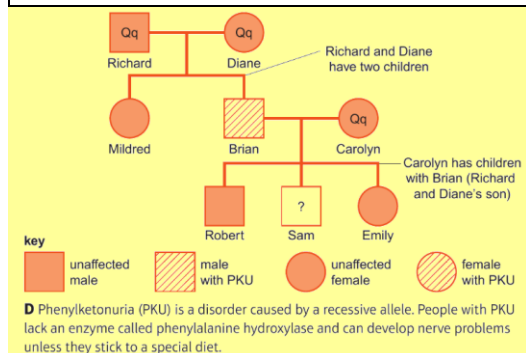
Prove it Questions

Page 31 Reproduction and Meiosis	
1. What is Meiosis and what does it produce?	(2)
2. How many Chromosomes are found within gametes?	(1)
3. Where in the body does Meiosis take place?	(2)
4. Compare the outcomes of Mitosis and Meiosis	(6)

Page 31 DNA	
1. State the shape of DNA	(1)
2. Identify what holds the bases together in DNA	(1)
3. Describe what is meant by "complementary base pairs)	(4)
4. Explain what a nucleotide is and why DNA is polymer?	(4)

Page 32 Extracting DNA	
1. List 4 stages in extracting DNA.	(2)
2. What breaks down the cell wall in the process of extracting DNA?	(1)
3. Describe how to release the DNA from its nuclear membrane.	(1)
4. Why do you need to add cold ethanol to the mixture when extracting DNA?	(2)

Page 32-34 Inheritance, Punnett squares	
1. What is an allele?	(1)
2. Blue eyes are represented by the letter b. If someone has the phenotype blue what is their genotype? And describe it in terms of homozygous/heterozygous recessive/dominant	(2)
3. Curly hair is represented by the allele r. One parent has the genotype Rr, another has the genotype rr. Calculate the probabilities of their children having curly hair. Use a diagram to help you.	(3)
4. The chances of two parents having a child with blue eyes was 50%, chances of them having a child with brown eyes was also 50%. They had 4 children – 3 blue and 1 brown eyed. How could this happen?	(3)



Page 34 Pedigree Analysis-	
Use the pedigree analysis chart below to answer these questions	
1. What letter represents the recessive allele?	(1)
2. Does Carolyn have PKU? What about her genotype tells you this?	(2)
3. Calculate the probabilities of Brian and Carolyn's children having PKU	(3)
4. Calculate the probabilities of Sam having PKU	(3)

Page 35 Variation	
1. What are the causes of genetic variation?	(2)
2. List 2 examples of genetic variation, environmental variation, and both	(3)
3. Data on foot length has been collected. Identify what type of data this is, why and how this should be represented.	(3)
4. What is normal distribution?	(3)

Page 36 Mutation	
1. What is a mutation?	(2)
2. What 3 things can cause mutations?	(3)
3. How can a mutation be positive (give an example)?	(3)
4. What effect can mutations have?	(3)

Mark scheme

Page 31 Reproduction and Meiosis	
1. Cell division (1) producing gametes. (1)	(2)
2. 23 (1)	(1)
3. Reproductive organs (1) Testes (1) Ovaries (1)	(2)
4. Mitosis – 1 cell division (1), Produces diploid cells (1) Creates identical copies (1) Meiosis 2 cell divisions (1) Produces haploid cells (1) Creates genetically different daughter cells (1)	(6)

Page 31 DNA	
1. Double helix (1)	(1)
2. (weak) hydrogen bonds (1)	(1)
3. Bases that always match to the same base. (1) For example Adenine – Thymine, (1) and Guanine to Cytosine. (1) 1 mark for naming all bases correctly.	(4)
4. A nucleotide is made of one sugar molecule, one phosphate molecule in the backbone (1) and one base (1) A polymer is a chain of repeating molecules (1) DNA is a polymer because it is a long chain of repeating molecules/nucleotides. (1)	(4)

Page 32 Extracting DNA	
1. Grind sample and mix with salt and detergent (1) Heat sample to 60°C for five minutes (1) Filter solution (1) Cool down and add ice cold ethanol (1)	(2)
2. Grinding/ crushing the sample. (1)	(1)
3. Mix crushed sample with salt and detergent (1)	(1)
4. To precipitate the DNA (1) from the filtered solution (1)	(2)

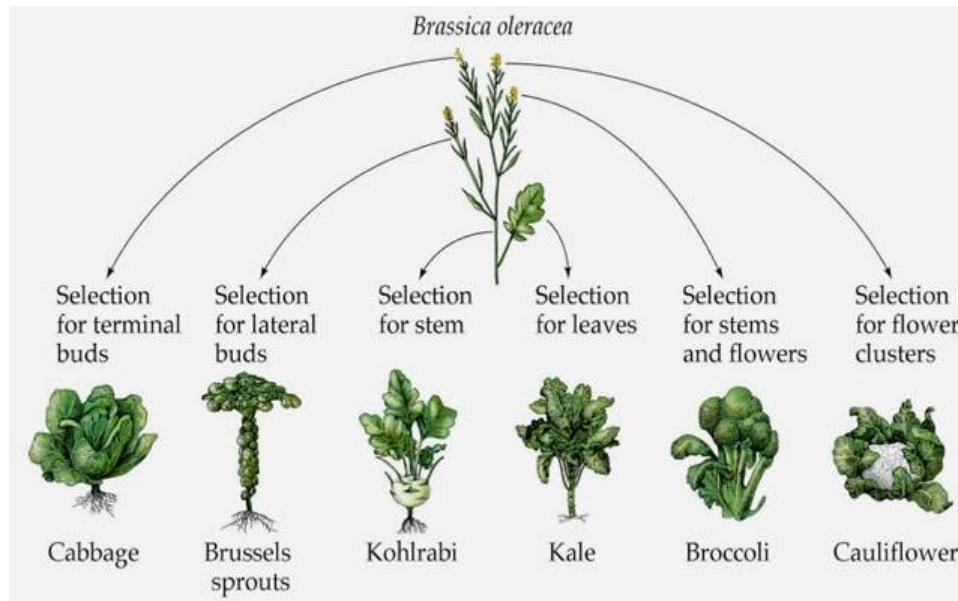
Page 32-34 Inheritance, punnett squares											
1. Two versions of the same gene. (1)		(1)									
2. bb (1) homozygous recessive. (1)		(2)									
3.		(3)									
	<table><tr><td></td><td>R</td><td>r</td></tr><tr><td>r</td><td>Rr</td><td>rr</td></tr><tr><td>r</td><td>Rr</td><td>rr</td></tr></table>		R	r	r	Rr	rr	r	Rr	rr	
	R	r									
r	Rr	rr									
r	Rr	rr									
50% having curly hair (rr)											
4. Punnett square would predict 50% blue 50% brown. (1) Actual offspring may not be 50% (1) The probability is applied to each child not the overall offspring (1)		(3)									

Page 34 Pedigree Analysis			
1. q			(1)
2. No (1) genotype Qq (1) she is a carrier of the recessive gene. (1)			(3)
3.			(3)
	q	q	
Q	Qq	Qq	
q	qq	qq	
50% chance of them suffering from PKU (1) 1 mark for correctly identifying Brians genotype as qq 1 mark for correctly identifying Carolyns genotype as Qq			
4.			(3)
	q	q	
Q	Qq	Qq	
q	qq	qq	
50% chance of Sam suffering from PKU (1) 2 marks awarded for recognition that probability for each child remains the same and the recognition that because 2 children are already known to not have PKU does not change the probability of Sam having PKU.			

Page 35 Variation		
1. Reproduction (1) Mutation (1)		(2)
2. Any correct two answers for each category award one point. genetic variation – Hair colour, eye colour, blood type, tongue rolling, genetic diseases (1) environmental variation – scars, accent, tattoos (1) both – Intelligence, Height, Weight (1)		(3)
3. This is continuous data (1) as it is quantitative (1) Histogram (1)		(3)
4. Data where majority of the sample is in a middle range, (1) and less of the sample is at either end. (1) Shows averages in a population sample (1)		(3)

Page 36 Mutation		
1. A mutation is a change in a gene or chromosome (1) A change in a base sequence (1) resulting in a changed phenotype (1)		(2)
2. UV Light, (1) Radiation, (1) Chemicals (1) Spontaneously (1)		(3)
3. Bacteria – Antibiotic resistance (1) Sickle cell Anaemia (1) Protected against malaria (1)		(3)
4. Change in a base sequence in DNA can lead to a change in a protein (1), which is either faulty or different to what is expected (1) The impact of this could mean that an important enzyme is not made. (1) Or a silent mutation where no effect is seen (1)		(3)

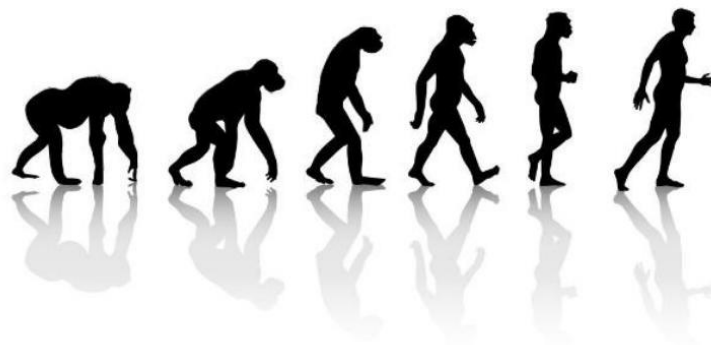
GCSE 9-1 BIOLOGY



Topic 4 – Natural Selection and Genetic Modification

Revision booklet

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Evidence for evolution	43-44
Classification	45
Selective Breeding	46
Genetic Engineering	46-47
Questions	48
Mark scheme	49-50

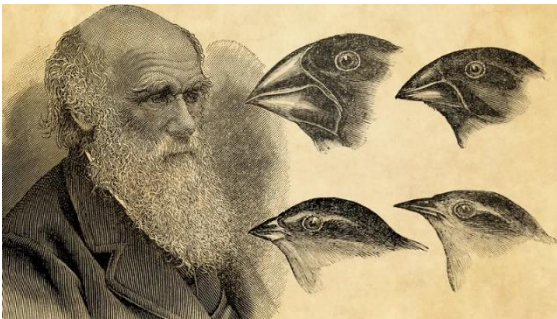


Darwin's theory of evolution

Evolution is the gradual and continual change of organisms over time. Charles Darwin is the scientist who came up with the theory of natural selection to explain how evolution occurred.

Darwin's theory had 5 major points:

- 1. Variation** – most populations of organisms contain individuals which vary slightly from one to another. The variation occurs because of mutation and sexual reproduction.
- 2. Competition** (Struggle for existence) – because populations do not generally increase rapidly in size there must therefore be considerable competition for survival between the organisms.
- 3. Survival** - those with advantageous characteristics are more likely to survive this struggle.
- 4. Reproduction** - Advantageous characteristics inherited – better adapted organisms are more likely to reproduce successfully passing on the advantageous characteristics to their offspring.
- 5. Gradual change** – over a period of time the proportion of individuals with the advantageous characteristics in the population will increase compared with the proportion of individuals with poorly adapted characteristics, and the poorly adapted characteristics may eventually be lost.



These points can be used to explain how over time a gradual change in the colour of the population of Cheetahs fur has changed, now appearing to be lighter than previously noted.

An alternative theory of evolution was proposed by Lamarck. He suggested organisms evolved due to needing certain characteristics that were acquired in response to the environment. For example, giraffes stretched their necks in order to get food from tall trees. He then stated that these

acquired characteristics were then passed onto their offspring. However genetic analysis has disproven this as we know that animals cannot change their genetic code in response to requiring a certain characteristic and therefore can't pass on these characteristics.

Evidence for evolution - Fossils

A fossil is the preserved remains of an organism, they can be found in rocks.

Fossil remains have been found in rocks of all ages. Fossils of the simplest organisms are found in the oldest rocks, and fossils of more complex organisms in the newest rocks. This supports Darwin's theory of evolution, which states that simple life forms gradually evolved into more complex ones.

There are gaps in the fossil record because many early forms of life were soft-bodied, which means that they have left few traces behind. What traces there were may have been destroyed by geological activity. This means Scientists cannot be certain how life began.

Famous fossils: Ardi – 4.4. million-year-old Human Ancestor

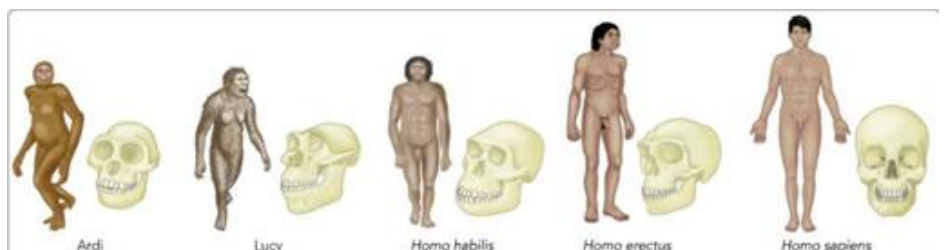
Features are a mixture of human and ape.

Ardi had a toe on the side of her foot suggesting she used to climb trees.

Long arms and short legs.

Brain size about the same as a chimpanzees.

Structure of legs suggested walked upright.



Famous fossils: Lucy – 3.2 million years old

Feet more suitable for walking than climbing (no toe on side of foot)

Size of arms and legs was between what you find in apes and humans.

Brain slightly larger than Ardi's but similar in size to a chimps brain.

Structure of leg bones and feet suggest better at walking upright than Ardi.

Leakey discovered human fossils from 1.6 million years ago. The more recent skeletons look more similar to humans than either Ardi or Lucy.

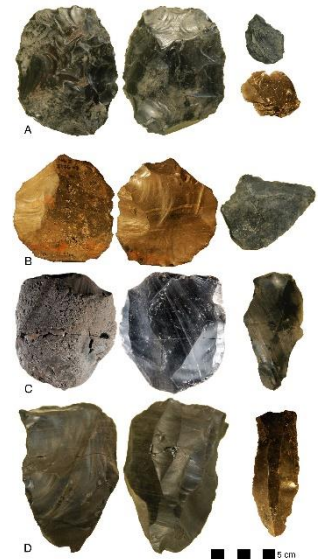
Scientists use this evidence of similarities between fossils to suggest links between the remains and believe this provides evidence for human evolution. However because there are gaps in the fossil record scientists can never be 100% certain that they are correct.

Evidence for evolution: Stone tools:

Uses of stone tools: Scrape meat from bones, hunting, building, weapons, digging, chopping etc. Scientists have also used stone tools to provide evidence for human evolution.

It was noted that stone tools have gradually become more complex over time. This has been linked to the volume of skulls found, as stone tools becoming more complex alongside the volume of human brains increasing. Suggesting that in order to make more complex tools our ancestors needed bigger brains that could think better than before. There are several ways to date stone tools, including simply looking at the features – simpler tools are more likely to be older than more complex tools. Other ways include the following:

1. **Radiometric carbon dating** - by looking at the natural radioactive decay of an **isotope** of Carbon (Carbon-14) we can estimate how long ago an organism lived. If any once-living material is found with a tool, such as a **piece of wood or fur**, we can date this to find the age of the rock.
2. **Stratifying rock layers** - looking at the layer of sediment in which a rock was found is a useful tool for archaeologists. Each layer of sediment, and everything within it, must have been formed at the same time. Therefore, we can date once-living **fossils** in this layer and use this to **estimate when the tools were formed**. By studying the rock layers they are found in. Older rock layers are normally found below younger layers. It can be better to date the rock the stone tools are found in as this lets us know when these stone tools were being used rather than when the rock that made them was formed.

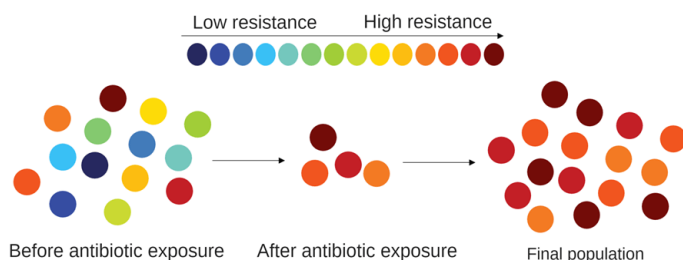


Evidence for evolution – Bacterial resistance

Over years bacteria has shown evolution by natural selection. Bacteria has become resistant to various antibiotics.

This can be explained by Darwin's theory of natural selection:

- Variation occurs because of mutation in the bacteria.
- Antibiotics are used to kill the bacteria.
- Antibiotics kills off most bacteria however the most resistant take the longest to die.
- Course of antibiotics is finished too soon leaving most resistant bacteria alive.
- This reduces competition allowing resistant bacteria to thrive and reproduce quickly.
- As bacteria reproduce asexually they produce genetically identical copies.
- The new population of bacteria are resistant to the antibiotics, as their offspring will be.



The new bacteria have been selected by the environment to have a feature (**resistance**) advantageous to survival. Therefore this is evidence of natural selection. As a result of this, population of antibiotic resistant bacteria increases. Bacterial diseases spreads rapidly because people are not immune to these new resistant bacteria and there is no treatment for it.

An example is MRSA. Called a 'superbug' as it is

resistant to many different types of antibiotics Common in hospitals: spreads when doctors and nurses move between patients.

Classification

5 Kingdoms versus 3 domains

It is important to classify organisms so that we know about the genetic relationships between species and organisms. We can do this by the old Five Kingdoms system or the newer Three Domains system.

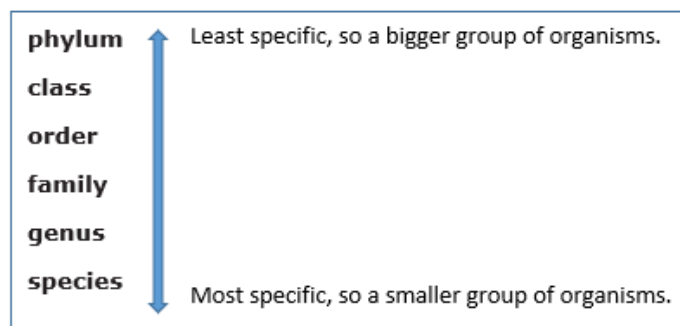
Five Kingdoms system

The Five Kingdoms Classification splits all organisms into one of 5 groups based on the features you can see in brackets:

- **Animals** (Multicellular, Heterotrophic)
- **Plants** (Multicellular, Autotrophic, Chloroplasts)
- **Fungi** (Multicellular, Saprophytic, Nucleus)
- **Prokaryotes** (e.g bacteria) (Unicellular, No nucleus)
- **Protists** (e.g algae, amoebas and other single-celled eukaryotic organisms) (Unicellular)

Each kingdom is then subdivided into the following taxonomy (in order from least specific to most specific)

These are different for each organism. For example, a human (*Homo sapiens*) would be the Animal kingdom, its phylum is Chordata, class is Mammalia and Order is Primate.



of

Binomial Naming System

The binomial naming system is based on the genus and species; for example, *Homo sapiens* is of the genus *Homo* and the species *sapien*,

Other organisms in the same genus but different families include *Homo habilis* and *Homo erectus*. The binomial naming system is important because it allows scientists to accurately identify individual species. For example, the European robin is *Erithacus rubecula*. It is much smaller than the American robin, *Turdus migratorius*, which belongs to a different genus.

Why change the classification system?

The above classification system is known as the Linnaeus system, this was based mainly on humans classifying things because of how they looked and observable characteristics. However with more scientific advances such as the improvement of the microscope and knowledge of DNA, we were able to examine organisms in more depth and this led to a change in how we classified organisms. The three Domain system is now used. We were able to compare DNA of different groups and recognised that Archaea have unused sections of DNA similar to Eukaryotes. This indicates they have evolved on a different pathway from Bacteria.

Three-domain system - Some species that we thought were originally very closely related are actually not. For example: it has been shown that the red panda and giant panda are not very closely related genetically. DNA analysis showed the giant panda to be a bear, and the red panda to be more closely related to a racoon.

So now before we split into kingdoms we have three bigger groups called Domains:

- **Archaea:** primitive bacteria which live in extreme environments such as hot springs. No Nuclei, unicellular, unused sections of DNA
- **Bacteria:** true bacteria (despite having similar features to archaea), No Nuclei, Unicellular. No unused sections of DNA.
- **Eukaryote:** organisms who have a nucleus enclosed in membranes, includes the kingdoms protists, fungi, plants and animals. Nuclei, Multicellular, unused sections of DNA.

Selective Breeding

Selective breeding is when humans choose which organisms to breed in order to produce offspring with a certain desirable characteristic (e.g animals with more meat, plants with disease resistance or big flowers). This has been happening for many years since animals were domesticated and plants were grown for food.

These are the stages of selective breeding:

1. Parents with desired characteristics are chosen.
2. They are bred together.
3. From the offspring those with desired characteristics are bred together.
4. The process is repeated many times until all the offspring have the desired characteristic.

Benefits of selective breeding	Disadvantages of selective breeding
It is possible to greatly increase the yield of a particular crop by selectively breeding only individuals that produce higher quality or a larger mass of food.	Selecting for advantageous characteristics can sometimes cause severe health problems in the offspring - e.g chickens that have been bred to have more meat (muscle) are sometimes too large to be able to walk.
Individual plants or animals can be bred to be resistant to a particular disease, which could increase crop yield.	Lack of genetic variation - Despite the bred population being able to have resistance to a particular disease (or multiple diseases), if one of them has susceptibility to a different disease then they all do - and the entire population could be wiped out as a result.
	There are ethical issues associated with selective breeding -many people consider it unethical to selectively breed for characteristics wanted by humans if it means that the offspring will suffer, or have a reduced quality of life as a result.

Genetic engineering

Genetic engineering: Modifying the genome of an organism by introducing a gene from another organism to give a desired characteristic.

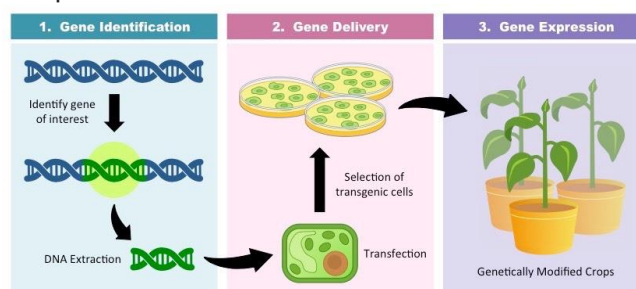
- Plant cells have been engineered for disease resistance or to have larger fruits.
- Bacterial cells have been engineered to produce substances useful to humans, such as human insulin to treat diabetes.

Genetically modified crops

- They are engineered to be resistant to insects and to herbicides.
- This will result in increased yields as less crops will die.

Genetic modification in medicine

- It may be possible to use genetic engineering to cure inherited disorders.
- It is called **gene therapy** and involves transferring normal genes (not faulty) into patients, so the correct proteins are produced.



Stages of Genetic Engineering (Higher only)

1. Genes from chromosomes are 'cut out' using restriction enzymes.
2. The same restriction enzymes are used to cut the vector (such as a virus or bacterial plasmid) into which the genes will be placed.
3. Ligase enzyme is used to attach the sticky ends of the gene and the vector together, to produce a recombinant gene product.
4. The vector is placed in another organism at an early stage in development so the desired gene moves into its cells and cause the organism to grow with the desired characteristics. In plants the vector is put into meristematic cells (unspecialised cells) which can then produce identical copies of the modified plant.

Advantages and Disadvantages of GM Organisms

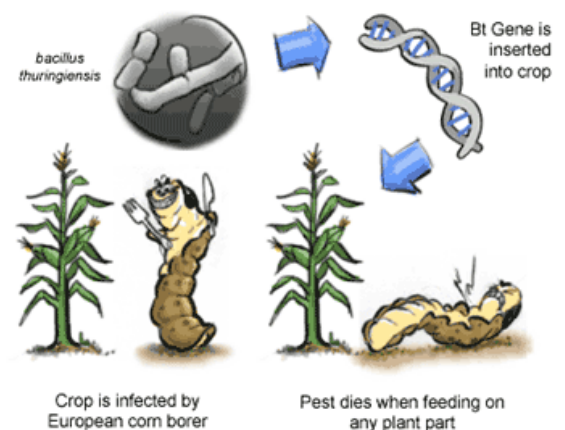
Benefits of Genetic Engineering	Risks of Genetic Engineering
It can be very useful in medicine to mass produce certain hormones in microorganisms (bacteria and fungi). Production of insulin etc.	GM crops might have an effect on wildflowers and therefore insects. • GM crops are infertile, and these genes could spread into wild plants, leading to infertility in other species, which affects the entire environment. • Growing with herbicides and pesticides can kill insects and other plants, which would reduce biodiversity.
In agriculture it can be used to improve yields by: • Improving growth rates • Introducing modifications that allow the crops to grow in different conditions, e.g. hotter or drier climates • Introducing modifications that allow plants to make their own pesticide or herbicide	People are worried that we do not completely understand the effects of GM crops on human health.
Crops with extra vitamins can be produced in areas where they are difficult to obtain. (Golden rice with extra beta carotene to help with vitamin A production and help with eyesight.)	Genetic engineering in agriculture could lead to genetic engineering in humans. This may result in people using the technology to have designer babies.
Greater yields can help solve world hunger, which is becoming an increasingly bigger issue due to population growth.	They pose a selection pressure, which could lead to increased resistance in other species, creating super weeds and pests.

BT crops

Bacillus thuringiensis is the name of a bacteria that produces toxins that kill insect larvae. This is a useful function for crops, so we use genes from the bacteria in crops to increase their insect resistance.

Genes are cut out from the bacteria using restriction enzymes, and re-inserted into the crop using ligase, (an enzyme). The crop will then produce the toxin - any insects that eat the crop will die.

As a result, less of the crop gets eaten by insects, increasing the crop yield and profits. However, there are concerns over this method - we don't know if the toxin has any effect on human health. Killing insects also results in a loss of biodiversity, which can affect the entire ecosystem.



Prove it Questions

Page 43 Darwin's Theory of Natural Selection	
1. What was Darwin famous for?	1
2. What causes variation in organisms?	2
3. Identify the 6 principles of Darwin's theory of natural selection?	6
4. It was noticed that a population of Elephants living in the Sahara Desert had begun to grow larger ears. This is an advantage in this area as the larger ears enable them to cool down quicker. Explain how this change in species could have occurred.	4

Page 43-44 Evidence for evolution	
1. Why can't scientists prove conclusively that fossils they have found are evidence of human evolution?	2
2. Why did scientists believe that Stone tools becoming more advanced over time was proof of evolution?	2
3. What evidence did Scientists use to link Ardi and Lucy?	2
4. Over the years some bacteria has become resistant to antibiotics, describe how this can be explained using Darwin's theory of natural selection.	6

Page 45 Classification	
1. List the taxonomy in order from least specific to most specific	2
2. Name the 5 kingdoms	2
3. Why have we recently been able to classify with greater detail resulting the development of the three domains	2
4. Name and describe the 3 domains.	3

Page 46 Selective breeding	
1. Suggest two characteristics a wheat breeder might want to select for	1
2. Suggest two characteristics a cattle breeder might select for.	1
3. A farmer would like to breed a goat that produces more milk. Describe what stages he would need to go through in order to this.	3
4. Describe two advantages and two disadvantages to selective breeding.	4

Page 46-47 Genetic Engineering	
1. Why have scientists only fairly recently been able to produce genetically modified organisms?	1
2. List two specific examples where genetic engineering has been used to benefit society.	2
3. Describe two advantages and two disadvantages of genetic engineering.	4
4. Compare Genetic engineering and selective breeding.	4
HIGHER ONLY	
5. Describe how scientists can use genetic engineering to manufacture insulin.	5

Mark scheme

Page 43 – Darwin’s Theory of Natural Selection	
1. Theory of natural selection to explain evolution.	1
2. Reproduction and mutation.	2
3. Variation, Overproduction, Competition, Survival of the fittest, Reproduction, gradual change.	6
4. Elephants showed variation in the size of ears, (1) This was an advantageous characteristic as it allowed them to cool down quicker, (1) therefore they were more likely to survive during difficult periods without shelter, (1) this led to elephants with this feature reproducing and passing on their advantageous characteristic to their offspring (1) Over time there was a gradual change in the species so the majority of the population showed this characteristic (1)	4

Page 43-44 Evidence for evolution	
1. There are too many gaps in the fossil evidence. (1)	2
2. Humans skull volume increased suggesting their brains size increased. Making advanced tools requires higher level cognitive processing (1) More advanced thoughts (1)	2
3. Similar bone structure (1) Both upright walkers (1) Same amount of fingers (1)	2
- Variation in the amount of resistance is shown in the bacteria. (1) - Antibiotics are used to kill the bacteria. (1) - Antibiotics kills off most bacteria however the most resistant take the longest to die. (1) - Course of antibiotics is finished too soon leaving most resistant bacteria alive. (1) - This reduces competition allowing resistant bacteria to thrive and reproduce quickly. (1) - As bacteria reproduce asexually they produce genetically identical copies. - The new population of bacteria are resistant to the antibiotics, as their offspring will be. (1)	6

Page 45 Classification	
1. Kingdom, Phylum, Class, order, Family, Genus, Species. For one mark you must have 3 in the correct order sequentially. For two marks you must have named all in the correct order. Full marks can be given if students miss out kingdom.	2
2. Animal, Plant, Fungi, Prokaryote, Protist. (3 correct for 1 mark, all correct for 2 marks)	2
3. Advances in Science, more specifically understanding DNA (1) Allows us to genetically classify organisms (1)	2
4. Bacteria, (cells with no nucleus, no unused sections in genes) (1) Eukarya, (cells with a nucleus, no unused sections in genes) (1) Archaea, (cells with no nucleus, genes contain unused sections of DNA) (1)	3

Page 46 Selective breeding	
1. Disease resistance (1) Yield – how much useful produce they might make (1) how well they cope with certain environmental conditions (1), fast growth (1), flavour (1)	1
2. Characteristics that allow them to cope with certain environmental conditions (1) Increased size – leading to increased yield (1) See above reasons.	1
3. Choose parents with desired characteristics (ability to produce more milk). (1) • Breed the parents together (1) • From the offspring pick those with desired characteristics and breed together. (1) • Repeat this process until all the offspring have the desired characteristic. (1)	3
4. Adv – Increase the yield (1) Breed organisms that are resistant to disease and therefore increase yield (1) Disadv – Severe health problems (1) Lack of variation could cause extinction if susceptible to certain diseases (1) Unethical to breed for characteristics to benefit humans if animal would suffer (1)	4

Page 46-47 Genetic Engineering	
1. Scientific advances in relation to understanding DNA and genetics as well as the techniques used to carry out genetic engineering are all very recent. (1) Human Genome project was only completed in 2003 (1)	1
2. Medicinal use – synthetic insulin to treat diabetes (1) Golden rice with more beta carotene to help eyesight in developing countries (1)	2
3. Adv – Medicine -mass produce certain hormones (1) Improve yields in agriculture (1) Produce crops with extra vitamins (1) Greater yields can help with food crisis due to increasing population (1) Disadv – GM crops could have a negative effect on wildflowers and therefore insects and reduce biodiversity. (1) Don't fully understand how GM crops affect human health (1) Agricultural genetic engineering could lead to genetic engineering in humans – designer babies and ethical issues (1) Selection pressure – increased resistance in other species (1)	4
4.	4
5.	5

GCSE 9-1 BIOLOGY



Topic 5 – Health, Disease and development of medicines.

Revision booklet

Topic	Pages
Health and Non-Communicable disease	52-53
Pathogens and Communicable diseases	54
STI's and Spreading pathogens	55
Body's defences	56
Immune system, Vaccinations and Antibiotics	57-59
Questions	60
Mark scheme	61-62



Health

Good health is not simply feeling well, it is a state of complete physical, social and mental wellbeing

Social well-being includes how well you get on with other people and how your surroundings affect you.

Mental well-being includes how you feel about yourself

Physical well-being includes being free from disease.

Communicable diseases are diseases that are caused by pathogens, and can be passed on in a variety of ways. For example, the Flu or Corona virus.

Non communicable diseases are diseases that cannot be passed on from person to person. They could be caused by genetics or they could be caused by lifestyle factors - such as drinking too much alcohol.

Diseases can be linked to one another so having one disease can be correlated with having another, if something damages your immune system it makes you more susceptible to getting another diseases. Periodontal disease is correlated with heart disease, as it causes bleeding in the gums, this allows pathogens to enter the blood and circulatory system.

Non-Communicable disease

Diseases that cannot be passed on from person to person.

There are two main types of non-communicable diseases, these include genetic disorders and deficiency disorders.

Examples of genetic disorders include type 1 diabetes, sickle cell anaemia, and cystic fibrosis. A genetic disorder is caused by faulty alleles, they can be passed onto offspring but not to anyone else.

Deficiency disorders are caused by malnutrition, the lack of specific nutrients can cause a specific deficiency disease:

Nutrient	Disease caused by deficiency of nutrient	Symptoms of disease	Good sources in diet
protein	kwashiorkor	enlarged belly, small muscles, failure to grow properly	meat, fish, dairy, eggs, pulses (e.g. lentils)
vitamin C	scurvy	swelling and bleeding gums, muscle and joint pain, tiredness	citrus fruits (e.g. oranges) and some vegetables (e.g. broccoli)
vitamin D and/or calcium	rickets or osteomalacia	soft bones, curved leg bones	vitamin D: oily fish calcium: dairy products
iron	anaemia	red blood cells that are smaller than normal and in reduced number, tiredness	red meat, dark green leafy vegetables, egg yolk

Alcohol and disease Some non-communicable diseases are caused by our lifestyle choices. An increased use of alcohol is linked to Liver disease, more specifically Cirrhosis of the liver. A Cirrhotic liver has a large amount of scar tissue which prevents it from working properly.

There is a positive correlation between amount of alcohol consumed and occurrence of Liver disease.

Cardiovascular disease

Malnutrition does not always mean that a person is not eating enough of certain nutrients, it can also mean a person is having too much of one thing such as sugars and fats. This can lead to obesity and a large amount of fat can be stored around the organs. Too much fat can increase the risk of cardiovascular disease.

We can calculate someone's BMI in order to determine whether or not they are obese.

$$\text{BMI} = \frac{\text{mass (kg)}}{\text{Height}^2 \text{ (m)}}$$

NB – do not forget to square the height. To square the height you multiply it by itself NOT BY 2. For example: if height is 1.5m
 $1.5 \times 1.5 = 2.25\text{m}$

For example: A person weighs 75kg and is 1.72m tall.

$$\text{BMI} = \frac{75}{1.72^2}$$

$$\text{BMI} = \frac{75}{2.96}$$

$$\text{BMI} = 25$$

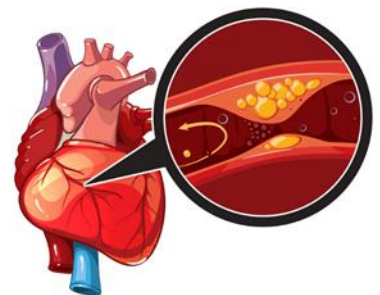
The BMI is used to determine if a person is overweight or not. If a person has a BMI of 30 or more they are usually considered to be obese. In the example above the BMI is 25 so this person is not considered to be obese.

A problem with using BMI to determine obesity is that it does not take into account muscle mass and the fact that muscle is denser than fat, so it is not the most accurate method to use.

Fat that seems to be more closely linked with cardiovascular disease is abdominal fat. Dividing waist measurement by hip measurement gives you a waist: hip ratio. This is a better method of measuring risk of cardiovascular disease in relation to someone's weight.

Smoking and disease

Substances found in cigarettes are toxic, they are linked to a number of health problems. One in particular is cardiovascular disease. Substances found within cigarettes damage the artery lining, which then allows fat to build up in the artery wall. This narrows the artery and prevents the easy flow of blood through this blood vessel. It could cause a blood clot which could then cause a heart attack or a stroke.



Treating Cardiovascular disease

Stent: this is a small mesh tube that can be inserted into a blocked blood vessel to hold it open allowing blood to flow through the vessel, this allows blood to transport Oxygen to the bodies organs.

Bypass surgery: arteries that are already blocked can be bypassed by inserting other blood vessels so blood flow can avoid the blocked arteries. Ensures that vital substances transported in the blood (Oxygen, Glucose and other nutrients) are able to reach the areas of the body where they are needed.

Both strategies are effective, but do not treat the symptoms. If lifestyle changes aren't made the problems will simply reoccur.

Pathogens and communicable diseases

There are 4 types of pathogen you need to be aware of:

- **Bacteria** – The cell structure of bacteria was covered in topic 1. These reproduce asexually so produce exact copies of parent cells.
- **Virus** – These are not technically classed as living organisms as they do not have a true cellular structure. They multiply by infecting a cell and taking over the cells DNA copying processes to make more viral. They cannot be treated with antibiotics as antibiotics don't target human cells so cannot affect a virus living inside a human cell.
- **Fungi** – Single celled or have a body made of hyphae, they can produce spores which can spread to other organisms.
- **Protists** – Some are parasitic so they live inside animals, the animals act as their hosts.

These pathogens are responsible for the transmission of communicable diseases.

We have a lot of bacteria that lives in our bodies but causes us no problems and is actually necessary in order for us to be healthy.

However, bacteria is also responsible for a number of diseases which can prove life threatening or at the very least inconvenient.

Common human diseases caused by pathogens:

Name of Disease	Category of Pathogen	Name of Pathogen	Effects	Method of Spread
Cholera	Bacteria	<i>Vibrio cholerae</i>	Diarrhoea	Water
Tuberculosis	Bacteria	<i>Mycobacterium tuberculosis</i>	Lung damage, coughing	Airborne
Chalara ash dieback	Fungi	<i>Hymenoscyphus fraxineus</i>	Leaf loss, bark lesions	Airborne
Malaria	Protists	<i>Plasmodium falciparum</i> (and others)	Damage to blood and liver	Animal vector (mosquito)
HIV	Virus	Human Immunodeficiency Virus	Destroys white blood cells, leads to onset of AIDS	Body fluids
Helicobacter	Bacteria	<i>Helicobacter pylori</i>	Can lead to stomach ulcers	Oral transmission
Ebola	Virus	<i>B. ebolavirus</i> (and others)	Causes hemorrhagic fever (fever accompanied by severe bleeding)	Body fluids

STI's

Name of STI	Category of Pathogen	Symptoms
Chlamydia	Bacteria	Often symptomless but if there are symptoms these can include painful urination or pelvic pain. Left untreated it can lead to infertility.
HIV	Virus	Increased susceptibility to other infections, severe illness and death if untreated

The spreading of pathogens can be reduced by using barrier methods of contraception such as condoms, or abstaining from sexual activity.

How are Pathogens spread?

Air – Coughing and sneezing spreads the pathogen in the air. A virus can survive for up to a day in the air. The bacteria that causes TB can survive for months by mingling with dust particles.

Water – Unclean water, and areas where there has been a war. Not often seen in developed countries due to the water being treated to kill pathogens. Cholera, dysentery and typhoid are passed through water.

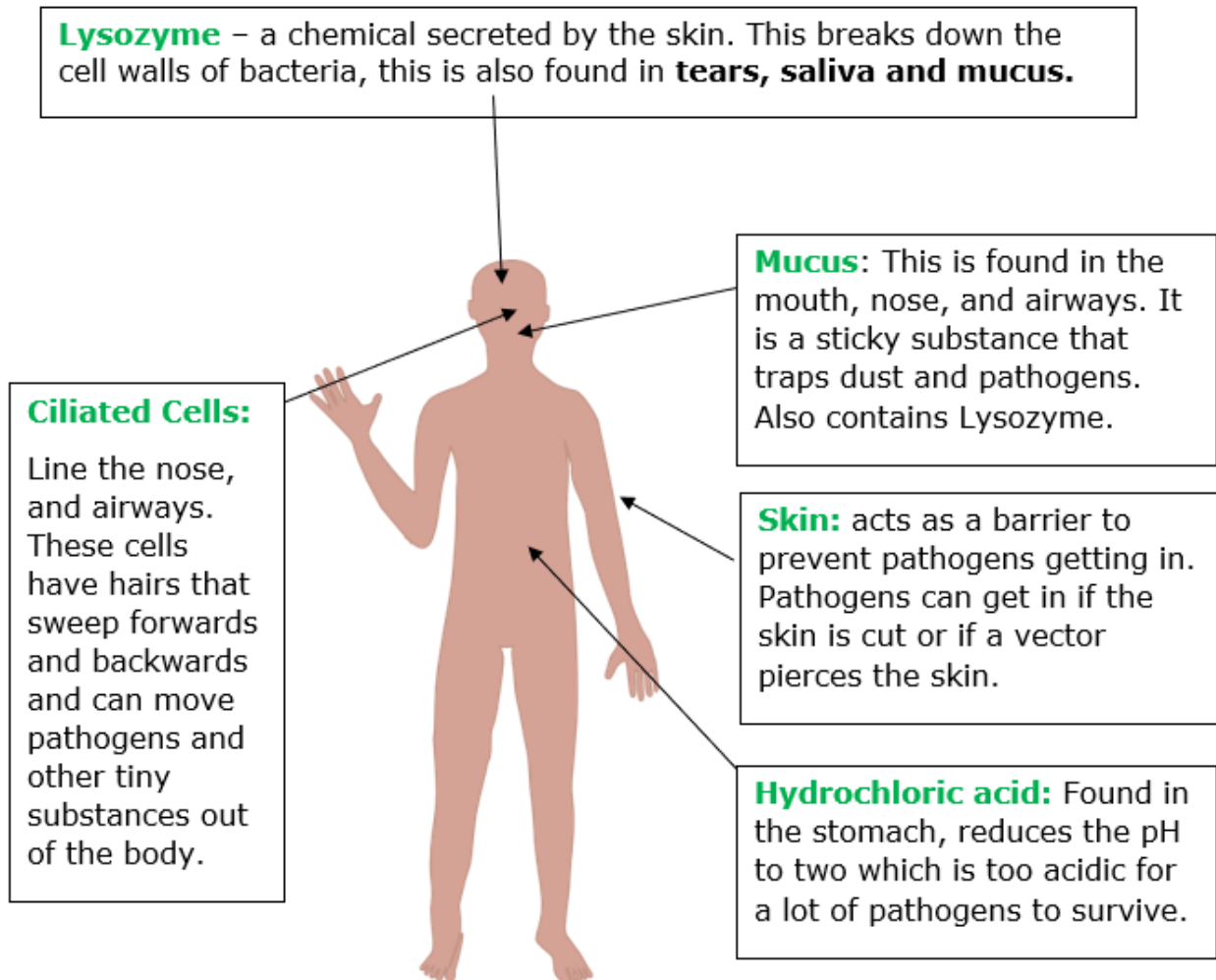
Food – contaminated food can enter the digestive system. This can happen if people handle food without washing their hands. Or if people do not wash their hands after going to the toilet etc. Also flies landing on food can cause food to become contaminated. This can cause food poisoning etc.

Vectors – Organisms can carry pathogens, for example Mosquitos carry the protest that causes malaria when it enters human blood.



The Body's defences

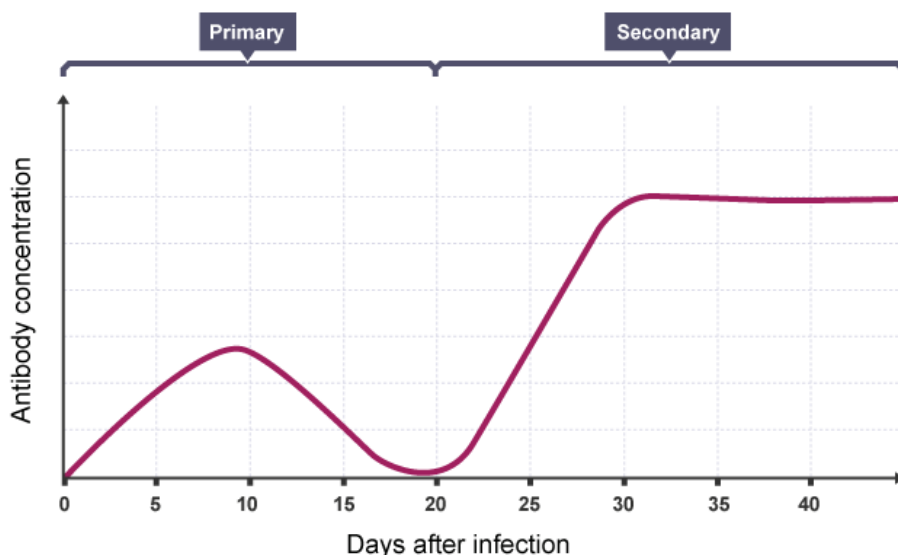
Pathogens can enter the body through any opening – including eyes, ears, nostrils, mouth, anus, vagina and any cut in your skin. To reduce the chances of pathogens entering in this way your body has a number of ways to prevent it.



The Immune System

If pathogens do enter your body, then your body will use its immune system to deal with them. Your body produces white blood cells in the bone marrow of your skeleton. White blood cells work to destroy pathogens in a variety of ways.

What happens	How it protects you
Phagocytosis – Phagocytes (one type of white blood cell, WBC) engulf and consume pathogens	This destroys the pathogens meaning they no longer make you feel ill.
Lymphocytes (WBC) Produce Antibodies.	Each pathogen has a chemical on their surface called an antigen, this has a specific structure that an antibody (produced by your lymphocytes) can bind to. When a lymphocyte is activated by an antigen it divides over and over again to make clones of identical lymphocytes. These secrete large amounts of antibodies that stick to the antigen and destroy the pathogen. When this happens the antigens also trigger production of memory lymphocytes. This means if you become infected again by the same pathogen the antibodies can be produced at a faster and more intense rate, meaning the individual will not feel the symptoms of the illness. They are said to be immune. This links to how vaccinations work.
Producing antitoxins	Neutralise toxins that are released by the pathogen.



This graph shows the immune response to a primary infection, and a secondary infection. The secondary infection occurs after the production of memory lymphocytes. This graph shows us the difference in immune response for primary and secondary, it demonstrates the increased production of antibodies and an increased speed in producing these for a secondary response in comparison to a primary response.

Vaccinations

Vaccinations are designed to make people immune to certain diseases. Discovered by Edward Jenner in 1796. By protecting the majority of the population the spread of the pathogen is reduced as there are less people to catch the disease from creating a situation known as "herd immunity". This is a situation the government hopes to create with the recent Corona Virus. Herd immunity protects more vulnerable members of society for whom certain illnesses pose a greater risk of fatality. If we can reduce the chances of the disease causing pathogens spreading then we protect vulnerable people. Vulnerable people include those with weaker immune systems due to a variety of reasons or elderly people.

How a vaccination works

1. Person is injected with a weakened or dead version of the microbe.
2. White blood cells produce antibodies to fight off the microbe.
3. If they get infected in the future the body will recognise the microbe and antibodies are produced quicker. (secondary response)
4. The microbe will be destroyed quickly so they don't get ill.

<u>Advantages of vaccination</u>	<u>Disadvantages of vaccination</u>
They have eradicated many diseases so far (e.g smallpox) and reduced the occurrence of many (e.g rubella).	They are not always effective in providing immunity.
Epidemics (lots of cases in an area) can be prevented through herd immunity.	Bad reactions (such as fevers) can occur in response to vaccines (although very rare).

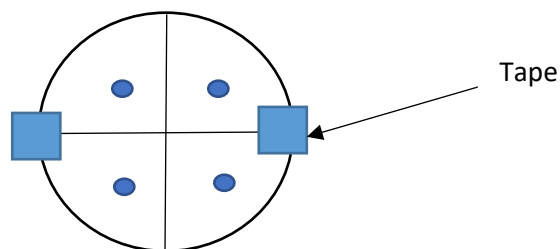
Antibiotics

Antibiotics can be used to treat diseases caused by bacteria. They cannot be used to treat diseases caused by viruses or by fungi. Antibiotics work because they inhibit cell processes in the bacteria cell. However, because viruses live in "host" cells and can only reproduce by using other cells machinery, antibiotics do not work.

You can investigate the effectiveness of antibiotics in the lab by using agar plates and antibiotics discs.

An agar plate is a petri dish that has been filled with agar jelly. Agar jelly is a substance that contains nutrients allowing bacteria to thrive and grow on them.

- Collect an Agar plate.
- Split the plate into 4 by drawing lines on the bottom of the agar plate.
- Label each section with the antibiotic you are going to place in each one.
- Place an antibiotic disc (paper containing antibiotic) into the matching labelled sections.
- Place the lid of the Agar plate and tape the lid down by putting a tab of tape on opposite sides of the plate.
- NB – DO NOT FULLY SEAL THE AGAR PLATE BY TAPING THE WHOLE WAY AROUND IT. This allows the growth of anaerobic bacteria that could be harmful.
- Incubate the bacteria for a period of a few days and then measure the distance between bacteria growth and the antibiotic. This is known as the zone of inhibition. The larger the distance the more effective the antibiotic against that specific bacteria.



Development of new drugs: Before drugs can be used to treat illnesses they have to go through rigorous testing procedures. You need to be able to describe these stages and give the reasons for each one of them.

1. The pre-clinical stage of testing is done on cells or tissues in the lab to find out whether the medicine gets into diseased cells and has the desired effect.
2. Testing may be carried out on animals to find out how the medicine affects the body systems, without risking human health.
3. A small clinical trial is carried out on a few healthy people to find out whether the medicine is safe for humans to take and that harmful side effects are limited.
4. A large clinical trial is carried out on many people with the disease to find out the right amount of medicine (dose) to use, and whether different people have different side effects.



Prove it Questions

<u>Page 52 - Health</u>	
1. Define Health.	1
2. What does social well-being mean?	1
3. What does mental well-being mean?	1
4. What does Physical well-being mean?	1

<u>Page 52-53 Non-Communicable disease</u>	
1. Define non-communicable disease	1
2. State two types of non-communicable disease	2
3. Describe two lifestyle factors that can lead to a non-communicable disease	2
4. Describe two ways that cardiovascular disease can be treated.	3

<u>Page 54-55 Pathogens, communicable disease, spreading pathogens</u>	
1. Name three types of pathogen.	1
2. Describe two diseases caused by bacteria.	2
3. Describe two diseases caused by a virus.	2
4. Describe symptoms of cholera, state what pathogen causes it and how it is spread.	4
5. Describe how malaria is transmitted.	3

<u>Page 56 The body's defences</u>	
1. What chemical is found on the skin and also in mucus, tears and saliva and what does it do?	2
2. Why can't a lot of pathogens survive in the stomach?	2
3. What does a ciliated cell do?	2
4. What substance lines the air ways and nostrils	2

<u>Page 57-58 The Immune system</u>	
1. What is phagocytosis	2
2. What role do memory lymphocytes play in the immune system?	2
3. What is the difference between a primary and a secondary response to an infection?	3
4. Describe how a vaccination works.	4

<u>Page 58-59 Antibiotics</u>	
1. What type of pathogen does antibiotics work to kill?	1
2. Why do antibiotics not work on viruses?	2
3. How do antibiotics kills bacteria?	1
4. Describe how you investigate the effectiveness of antibiotics.	5

Mark scheme

Page 52 - Health	
1. A state of complete social, physical and mental well-being. (1)	1
2. How well you can communicate with others around you, get on with others, and how your surroundings effect you. (1)	1
3. How good you feel about yourself (1)	1
4. Being free from disease (1)	1

Page 52-53 Non-Communicable disease	
1. A disease that cannot be passed onto others (1) A disease caused by lifestyle factors or genetics (1)	1
2. Nutrition deficiency (1) Genetic (1) Lifestyle (1)	2
3. Alcohol - Increased use of alcohol use can cause liver damage (cirrhosis) Smoking – Damages arteries causing blockages and poor blood flow, leading to possible strokes or heart attacks (1) Poor diet – too much fat building up around the organs, could cause blockages in arteries preventing blood flow, cause heart attacks, strokes (1)	2
4. Stent – Insert a small mesh tube to hold a blood vessel open and allow blood to flow freely (1) Heart bypass – insert other blood vessels to bypass ones that are blocked allowing blood to flow freely and transport other nutrients (1) 1 mark allocated for identification of two treatments. The remaining two marks come from the descriptions.	3

Page 54-55 Pathogens, communicable disease, spreading pathogens	
1. Bacteria, Fungi, Virus, Protist (any three for one mark)	1
2. Cholera – causes diarrhoea, passed in water. (1) Tuberculosis causes lung damage and is airborne. (1) Helicobacter – causes food poisoning, stomach ulcers. Passed orally (1)	2
3. Influenza, causes cold like symptoms airborne (1) HIV – destroys white blood cells leads to AIDS, transmitted through bodily fluids. (1) Ebola cause severe fever and internal bleeding transmitted through bodily fluids (1). Could also include cold, Corona virus both of these are airborne.	2
4. Cholera causes severe diarrhoea. (1) It is transmitted through water (1) and caused by bacteria. (1) Not often seen in developed countries where water is routinely treated to kill pathogens.	3
5. Mosquito acts a vector for the protist that causes malaria (1) When mosquito pierces skin to drink blood (1) they inject the protist into the blood stream (1)	3

Page 56 The body's defences	
1. Lysozyme. (1) Breaks cell wall of bacteria down (1)	2
2. Hydrochloric acid in stomach (1) reduce the pH to 2 (1) too acidic for majority of pathogens to survive (1)	2
3. Found in air ways and nostrils (1) Hairs on ciliated cell (1) move forwards and backwards to trap dust and pathogens (1) and sweep them out of body (1)	2
4. Mucus (1)	1

Page 57-58 The Immune system	
1. When white blood cells(phagocytes) (1) engulf and digest pathogens in the body (1)	2
2. Produced when body has encountered pathogen and have specific antibodies (1) When secondary infection occurs memory lymphocytes respond quickly to prevent person feeling unwell (1)	2
3. Primary – Lymphocytes produce antibodies to deal specifically with antigens o pathogen to get rid of pathogen (1) Takes a longer time (1) Secondary response relies on memory lymphocytes producing antibodies (1) Faster response (1) What is the difference between a primary and a secondary response to an infection?	3
• Person is injected with a weakened or dead version of the microbe. • White blood cells produce antibodies to fight off the microbe. • If they get infected in the future the body will recognise the microbe and antibodies are produced quicker. (Secondary response) • The microbe will be destroyed quickly so they don't get ill.	4

Page 58 Antibiotics	
1. Bacteria (1)	1
2. Antibiotics inhibit cell processes in the bacterial cell. As viruses live in host cells they cannot do this to a virus (1)	2
3. Stop /inhibit cell processes from happening. (1)	1
• Collect an Agar plate. • Split the plate into 4 by drawing lines on the bottom of the agar plate. (1) • Label each section with the antibiotic you are going to place in each one. (1) • Place an antibiotic disc (paper containing antibiotic) into the matching labelled sections. (1) • Place the lid of the Agar plate and tape the lid down by putting a tab of tape on opposite sides of the plate. (1) • Incubate the bacteria for a period of a few days and then measure the distance between bacteria growth and the antibiotic. This is known as the zone of inhibition. The larger the distance the more effective the antibiotic against that specific bacteria. (1)	5