

LIFE SCIENCES

Grade 12 Textbook



basic education

Department:
Basic Education
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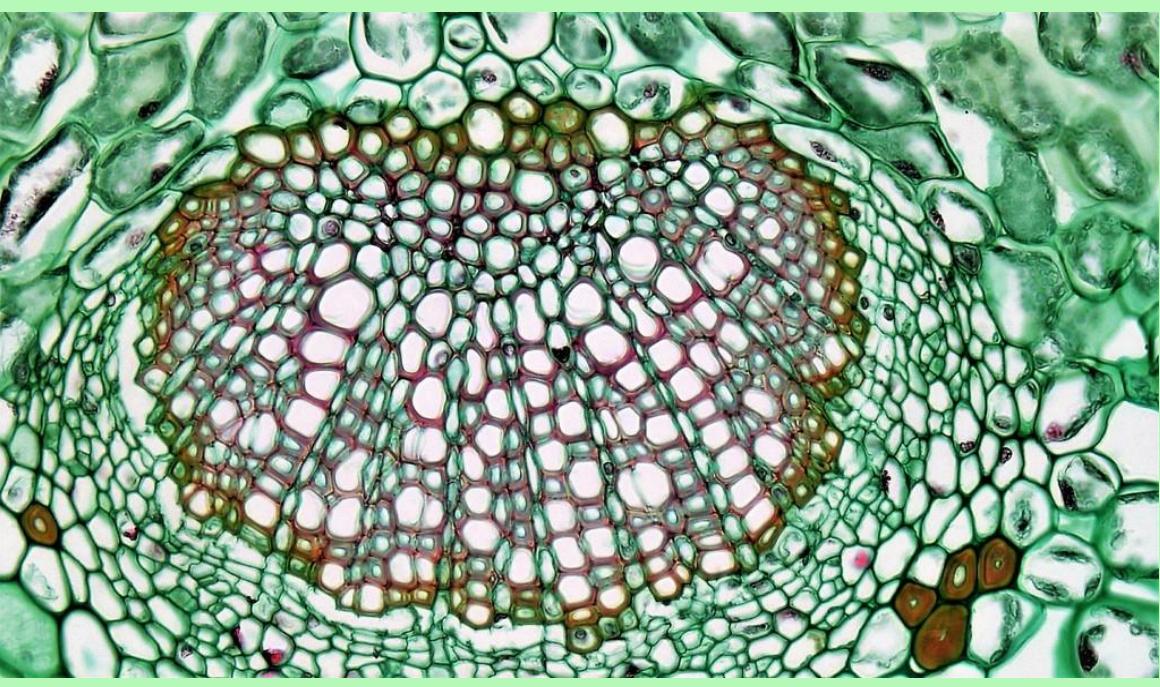
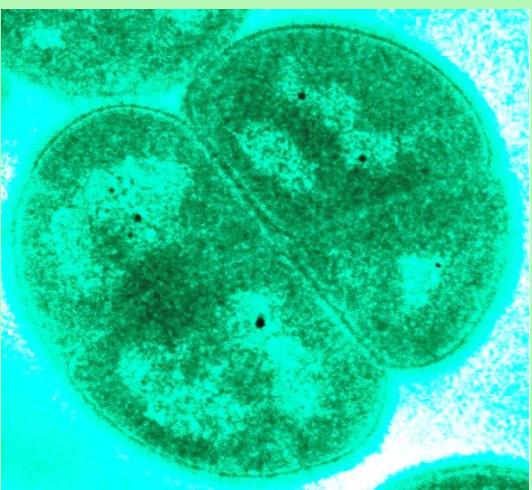
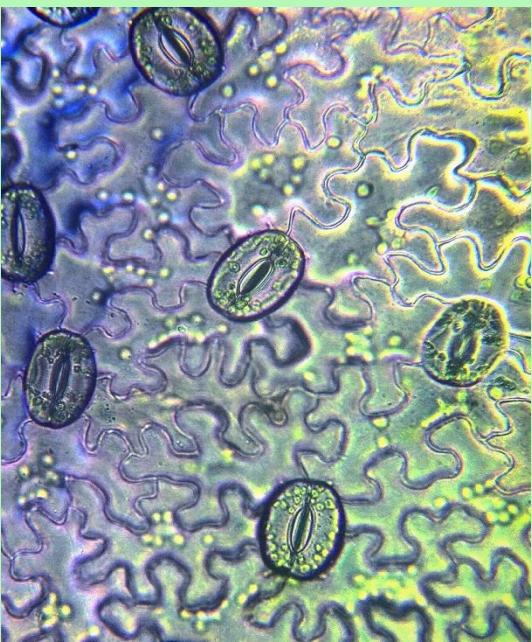
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INTRODUCING LIFE SCIENCES

The aim of this textbook is to allow you, the learner, to be an active partner in your learning experience. The text has been designed to cover all the content you need for Grade 12, and to provide it in a readable manner that communicates all concepts simply, clearly and in the necessary amount of detail.

The next few pages will provide you with a broad overview of Life Sciences and hopefully show you its value as a choice for a school subject.

Studying Life Sciences also offers you broader benefits: it will encourage your ability to **think critically**, to **solve problems** and seek to **understand the world around you**.

What are the Life Sciences?

The term 'Life Sciences' indicates clearly the two ideas held together in this subject:

- **Life** refers to all living things – from the most basic of molecules through to the interactions of organisms with one another and their environments.
- **Science** indicates it is necessary to use certain methods in our study of the subject. The two broad aims of any science are to increase existing knowledge and discover new things.

We approach this using careful methods that can be copied by others. These include:

- proposing hypotheses (the predicted outcome of an investigation) and
- carrying out investigations and experiments to test these hypotheses.

Scientific knowledge changes over time as more is discovered and understood about our world; as such, Life Sciences is a constantly growing subject.

Why choose Life Sciences as a subject?

- **First**, to give you knowledge and skills that are helpful in everyday life, even if you do not pursue Life Sciences after school.
- **Secondly**, to expose you to the wide variety of sub-fields within the subject that could encourage or interest you to pursue a career in the sciences.

If you choose to study Life Sciences at school you will be able to study any Life Science specialisations after school - such as microbiology, genetics, environmental studies or biotechnology.

What skills will Life Sciences equip you with?

This subject will teach you important biological concepts, processes, systems and theories, and provide you with the skills to think, read and write about them. Life Sciences will:

- give you the ability to evaluate and discuss scientific issues and processes
- provide an awareness of the ways biotechnology and a knowledge of Life Sciences have benefited humankind
- show you the ways in which humans have impacted negatively on the environment and organisms within it, and show you how to be a responsible citizen in terms of the environment and conservation
- build an appreciation of the unique contribution of South Africa to Life Sciences - both the diversity of the unique biomes within Southern Africa and the contributions of South Africans to the scientific landscape.

Life Sciences Strands for Grade 12

Everything you study this year will fit into one of these three broad strands. These knowledge pathways grow over your three years of FET.

Within each knowledge strand, ideas should not be studied separately; rather seek to discover the links between related topics so that you grow in your understanding of the inter-connectedness of life. As you study each section or chapter, look for the broad strokes that place it under one of these strands:

- Knowledge **Strand 1**: Life at a Molecular, Cellular and Tissue Level
- Knowledge **Strand 2**: Life Processes in Plants and Animals
- Knowledge **Strand 3**: Diversity, Change and Continuity

The Purpose of studying Life Sciences

There are three broad purposes, which will expand as we continue:

- **Aim 1** - knowing the content (theory);
- **Aim 2** - doing practical work and investigations;
- **Aim 3** - understanding the applications of Life Sciences in society - both present society (indigenous and western) and within the context of history.

Aim 1: Knowing the content of Life Sciences

Learning content involves understanding and making meaning of scientific ideas, and then connecting these ideas. Theory is not just recalling facts; it is being able to *select important ideas, use different sources to learn, and describe concepts*,

processes and theories important to Life Sciences.

Within this you will learn to *write summaries*, develop your own *diagrams* and *reorganise data* you are given into something meaningful. Additionally, you will learn to *interpret the data* you are working with and *link it to theory* you have studied.

Aim 2: Doing practical work and investigations

Life Science is a fascinating subject and one of the best ways to understand it is for you to see it in action. Therefore, it is important for you to know how to do practical investigations. Within this, you will learn many useful skills like how to *follow instructions* in a safe manner and how to *name, recognise and handle laboratory equipment*.

During a practical investigation it is important for you to be able to *make observations*. There are many ways this can be done - by making drawings, describing what you see, taking measurements, and comparing materials before and after a certain treatment. After making these observations it is important for you to be able to *measure and record them* in a useful way. From here you will *interpret your data* - you will look for the value in what you gathered and discuss the changes, trends, and applications of what you have shown.

Finally, you will learn how to design your own investigations and experiments. An investigation is more straightforward; for example, it could involve observing soil profiles or counting animal populations.

Planning an experiment would begin with identifying a problem, and then hypothesising a solution. In planning, you would identify variables and consider ways to control them, select apparatus and materials to assist you, and then plan an experiment that could be repeated by someone else. It is also important to consider ways of capturing and interpreting your data.

Aim 3: Understanding the history, importance and modern applications of Life sciences

The third aim of Life Sciences is to show you that school science can be relevant to your life and that studying provides enrichment to you, even if you do not pursue it past school level.

As you study you will be exposed to the history of science and indigenous knowledge systems from other times and other cultures. As you learn a certain section of work, you will be introduced to how that knowledge was developed by various scientists across the ages as they pursued a deeper understanding of the world around them.

Our search of knowledge is shaped by our world view. Therefore, an important concept to be aware of is that modern science (and technology) and traditional, indigenous knowledge systems will sometimes differ in their approach to science.

These seemingly opposite views can be held together as both bring a certain dynamic; they should not be seen as opposing forces.

Finally, there are many possible career fields branching out of Life Sciences and, as you learn, some of these will be shown through examples. Different sections would open up different careers choices- *in the past* (for example palaeontology), *the present* (like horticulture, game ranch management and preservation) and *the future* (such as biotechnology and genetic engineering).

A final word on using this textbook

The best way to use this textbook to increase your understanding and thereby results would be as follows:

- Remember, good learning begins in the classroom so always pay careful attention as your teacher works through it with you
- Take note of sections you do not understand and revisit them
- Ask questions to make sure you understand
- Consider the end-of-chapter summaries and build on them to create your own point-form summary note
- Practice re-sketching the given diagrams
- Work through all the given questions and answers at the end of the chapter

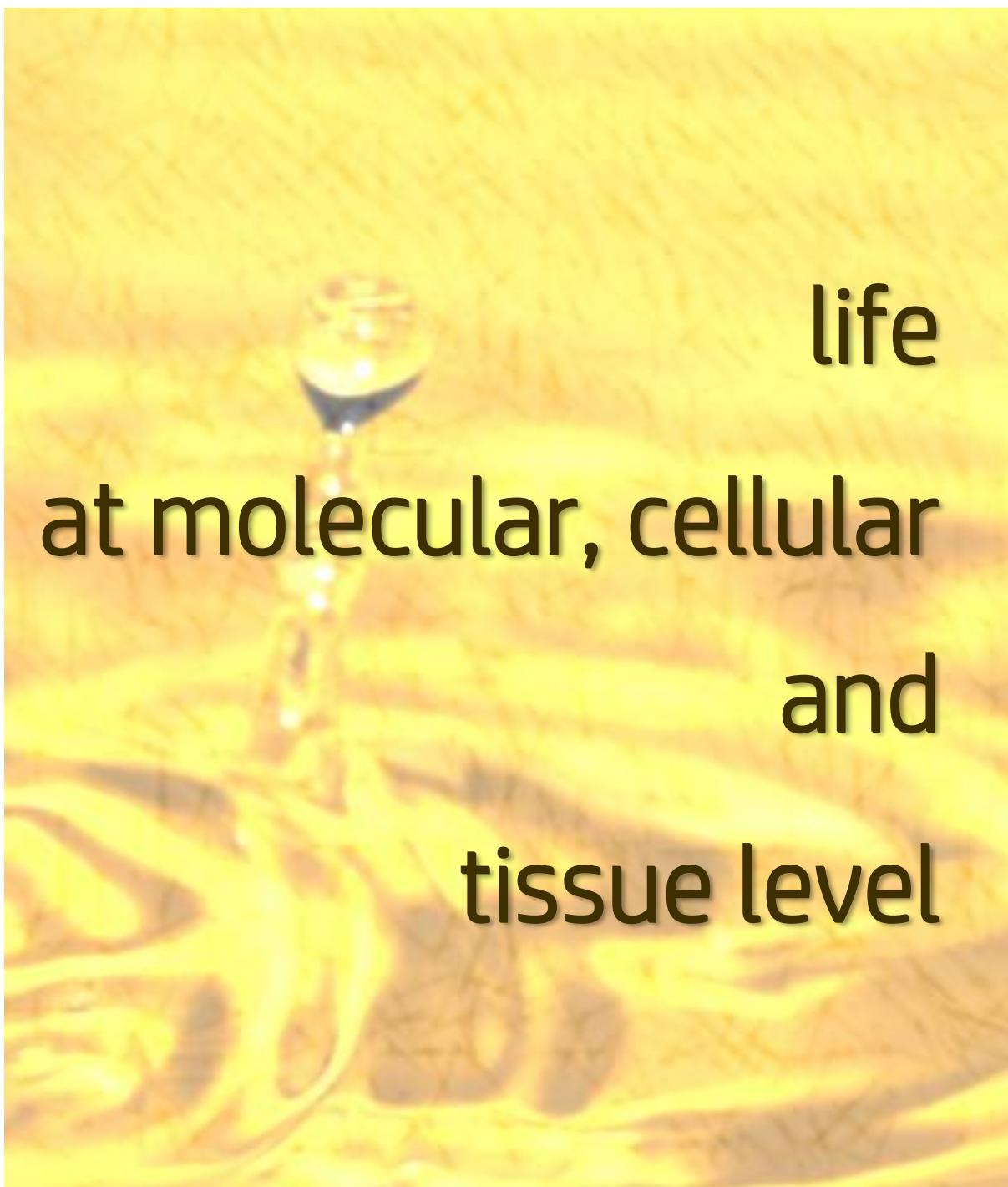
Strand

life

at molecular, cellular

and

tissue level



1: DNA – the code of life

Introduction	Activity 2: DNA replication
Revision of cellular structure	DNA profiling
The structure of nucleic acids	Activity 3: DNA profiling
DNA – deoxyribonucleic acid	Protein synthesis
A brief history of the discovery of DNA	Protein synthesis occurs in two stages
The location of DNA	Stage 1: Transcription
The structure of DNA	Stage 2: Translation
The role of DNA	The effect of mutation on protein structure (DNA sequence)
Activity 1: DNA	Activity 4: Protein synthesis
RNA – ribonucleic acid	Activity 5: Codons and amino acids
The location of RNA	
The structure of RNA	
The role of RNA	
Comparison between DNA and RNA	End of topic exercises
DNA replication	
Errors that occur during DNA replication	

CHAPTER 1: DNA – THE CODE OF LIFE

Introduction

- All living organisms contain both DNA (deoxyribonucleic acid) and RNA (ribonucleic acid) – we focus on their location, structure and function.
- We explore the discovery of DNA, its role in the human body and how it replicates.
- Protein synthesis is vital for life – we examine how proteins are formed by both DNA and RNA.

Revision of cellular structure

It is important to know the location and functions of certain organelles, illustrated in Figure 1 below.

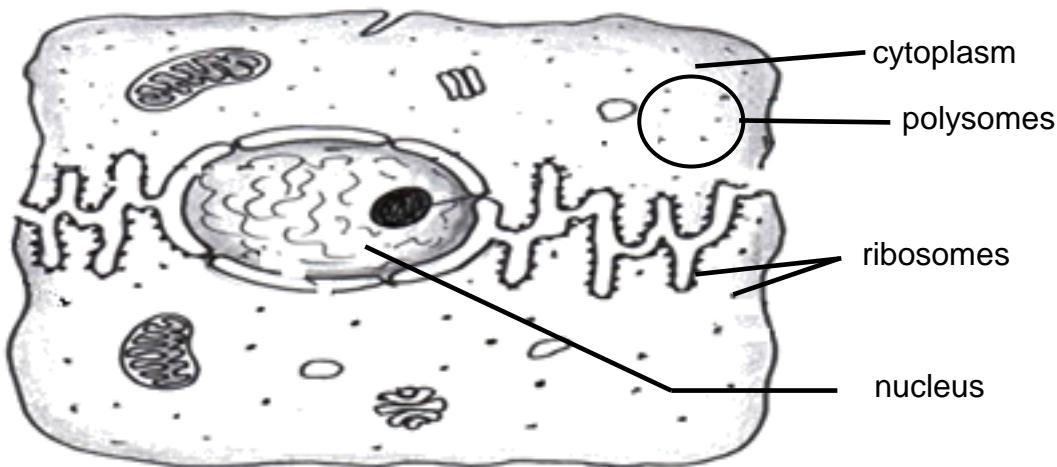


Figure 1: Structure of a cell

Cytoplasm is the base substance in which the organelles of the cell are suspended. It is a watery substance and allows for metabolic reactions to take place.

Ribosomes are small, round organelles which are mainly found attached to the endoplasmic reticulum or are free-floating in the cytoplasm. Ribosomes can also be found inside other organelles such as the chloroplast and mitochondria but in smaller numbers. They are the site of protein synthesis and consist of RNA and protein.

The **nucleus** controls all of the cell's activities.

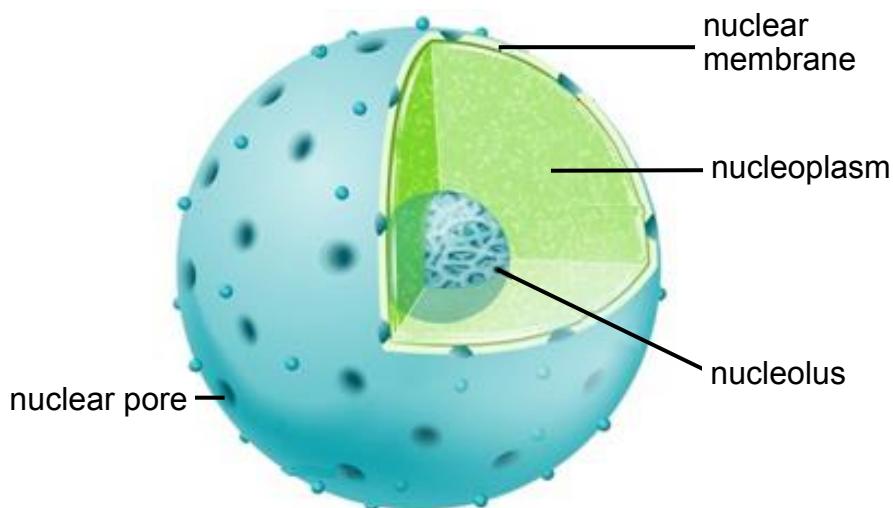


Figure 2: Parts of the nucleus

A nucleus has four main parts:

- the double **nuclear membrane** – it encloses the nucleus and contains small pores to allow for the passage of substances in and out of the nucleus
- the **nucleoplasm** – this is a jelly-like fluid within the nucleus
- the **nucleolus** – a dark body suspended in the nucleoplasm which contains free nucleotide bases and produces ribosomes
- the **chromatin network** – found in the nucleoplasm: contains the DNA which forms the chromosomes containing the genetic code of a person / organism

The structure of nucleic acids

Key terminology

nucleic acid	a type of organic compound
monomer	a building block
nucleotide	the monomer which forms DNA and RNA

There are two types of nucleic acids in the human body – DNA (deoxyribonucleic acid) and RNA (ribonucleic acid). Together these form the basis of all life on earth. They consist of monomers (building blocks) called nucleotides.

The basic structure of a nucleotide is illustrated in Figure 3 below. Each nucleic acid is composed of a phosphate group (P), a sugar molecule (S) and a nitrogenous base (NB).

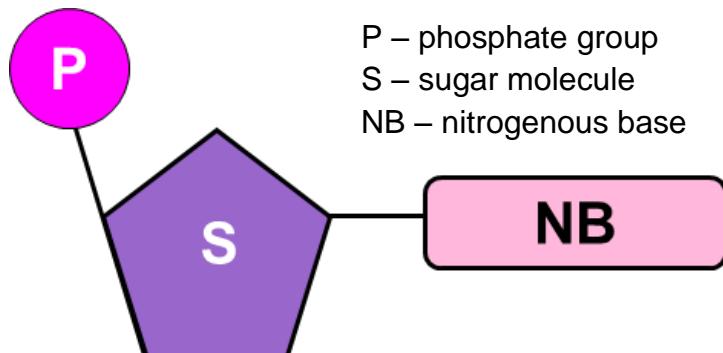


Figure 3: A nucleotide

DNA – deoxyribonucleic acid

Key terminology

DNA	<ul style="list-style-type: none">deoxyribonucleic acid is made up of nucleotidesnitrogenous bases adenine, thymine, guanine and cytosinecarries the genetic code for protein synthesis
nuclear DNA	DNA found in the nucleus
extra- nuclear DNA	DNA found outside of the nucleus: mitochondrial and chloroplastic DNA.
double helix	the shape of DNA consists of two strands joined together and twisted spirally
hereditary	genetic information passed on from parent to offspring

A brief history of the discovery of DNA

- 1952 – Rosalind Franklin and her assistant Maurice Wilkins researched the structure of DNA using X-ray diffraction images.
- Watson and Crick did independent research on DNA. Upon seeing Franklin's images, they proposed a 3-D double helix model for DNA in 1953.
- 1962 – Watson and Crick received the Nobel Prize for the discovery of the structure of DNA, and Wilkins received an award for his X-ray photography. Franklin had died of cancer.

Rosalind Franklin – background: <https://www.youtube.com/watch?v=BIP0IYrdrl>

The location of DNA

DNA is found in **two locations** in a cell:

- Mostly in the nucleus of a cell – this is referred to as **nuclear DNA**
- a small amount is found outside the nucleus – it is referred to as **extra-nuclear DNA**. There are two types of extra-nuclear DNA:
 - chloroplastic DNA – found in the chloroplasts of plant cells
 - mitochondrial DNA – found in the mitochondria (useful for tracing ancestry)

The structure of DNA

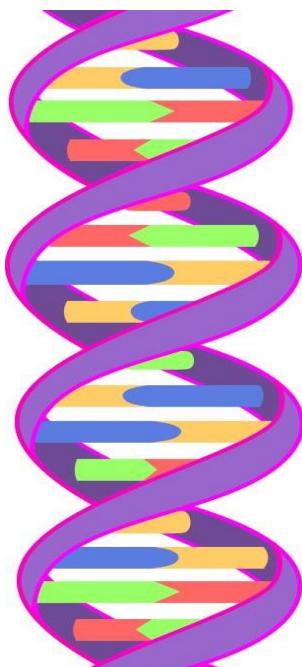


Figure 4A: DNA double helix

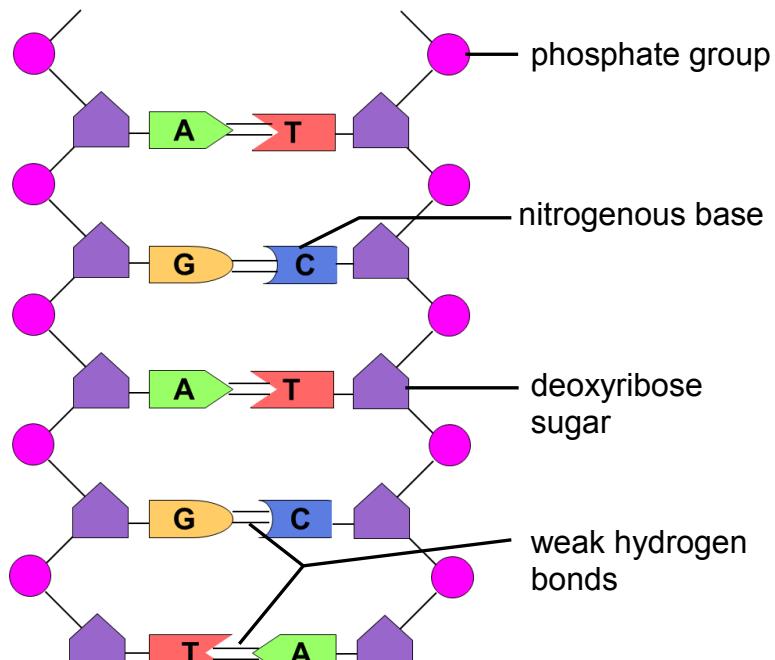


Figure 4B: DNA – simplified structure

DNA has a double helix structure (Figure 4A), consisting of monomers called nucleotides which link to form long chains, called polymers. The sugar in DNA is deoxyribose sugar and is attached to a nitrogenous base. The phosphate and sugar molecules are attached to one another by strong bonds alternately to form the long chains (Figure 4B).

There are four types of nitrogenous bases in DNA:

adenine (A)

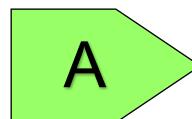
cytosine (C)

thymine (T)

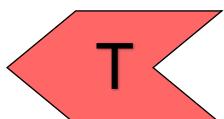
guanine (G)

Nitrogenous bases are complementary, and always join together in a specific order:

- adenine always links to thymine (Figure 5A)
- guanine always links with cytosine (Figure 5B)



adenine



thymine



guanine



cytosine

Figure 5A: adenine with thymine

Figure 5B: guanine with cytosine

This pairing of bases means that two strands of DNA are joined together, forming a long ladder-like structure. The nitrogenous bases are held together by weak hydrogen bonds. The ladder-like structure becomes coiled and is known as a **double helix structure**. The DNA strands wind around proteins which are known as histones.

The role of DNA

DNA carries hereditary information in the form of genes. Genes are short sections of DNA which code for a specific trait, and determine the physical characteristics (e.g. blood grouping, a gene linked to breast cancer) and behaviour of an organism (e.g. whether an organism can be tamed and domesticated).

Most of the DNA strands do not code for anything and are known as non-coding DNA. Scientists are still researching the importance of the non-coding DNA.

The main functions of DNA include:

- Controls the functioning of cells
- Regulate the functioning of genes
- Passes on hereditary characteristics

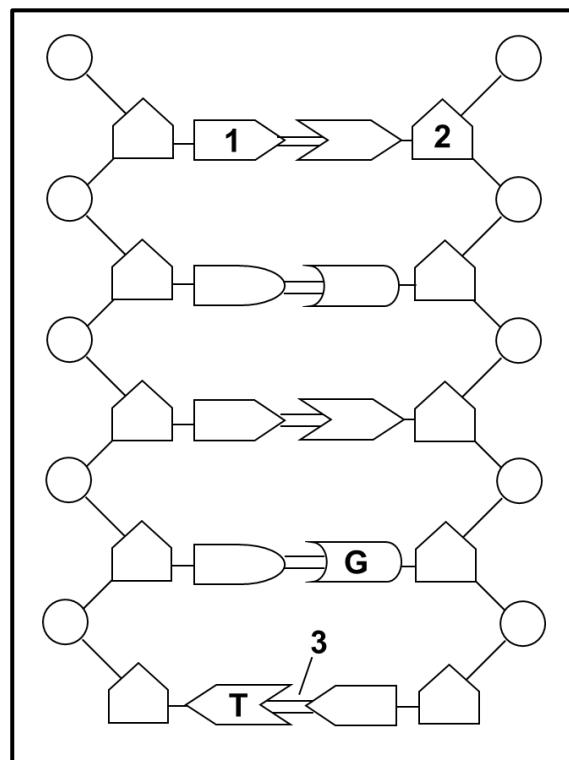
The structure of DNA: <https://www.youtube.com/watch?v=C1CRtkWwu0>

Activity 1: DNA

The diagram on the next page shows part of a DNA molecule.

1. Label parts 1, 2 and 3
2. Give the number of nucleotides shown in the diagram
3. Name two places in an animal cell where this nucleic acid may be found.
4. What is the natural shape of this molecule?
5. Draw a nucleotide with the nitrogenous base adenine.

(3)
(1)
(2)
(1)
(4)
(11)



RNA – ribonucleic acid

Key terminology

RNA	RNA consists of nucleotides. Nitrogenous bases found in RNA are adenine, uracil, guanine and cytosine
messenger RNA	mRNA carries the code for protein synthesis from DNA to the ribosome
ribosomal RNA	rRNA forms ribosomes which are the site of protein synthesis
transfer RNA	tRNA brings amino acids to the ribosome to form the protein

There are three types of RNA (ribonucleic acid), all formed in the nucleus by DNA. They perform different functions in different places in a cell. The types are:

- messenger RNA (mRNA)
- ribosomal RNA (rRNA)
- transfer RNA (tRNA)

The location of RNA

- **Messenger RNA (mRNA)** is formed in the nucleus but then enters the cytoplasm where it attaches to ribosomes.
- **Ribosomal RNA (rRNA)** is found in the ribosomes in the cytoplasm of the cell.
- **Transfer RNA (tRNA)** is found freely in the cytoplasm of the cell.

The structure of RNA

Like DNA, RNA also consists of monomers (nucleotides) which link to form longer chains (polymers).

However, RNA is a **single-stranded structure** which is not coiled. The sugar in RNA is ribose and is attached to a nitrogenous base. The phosphate and sugar molecules are attached to one another alternately to form the chains.

The structure of RNA is illustrated in Figure 6 below.

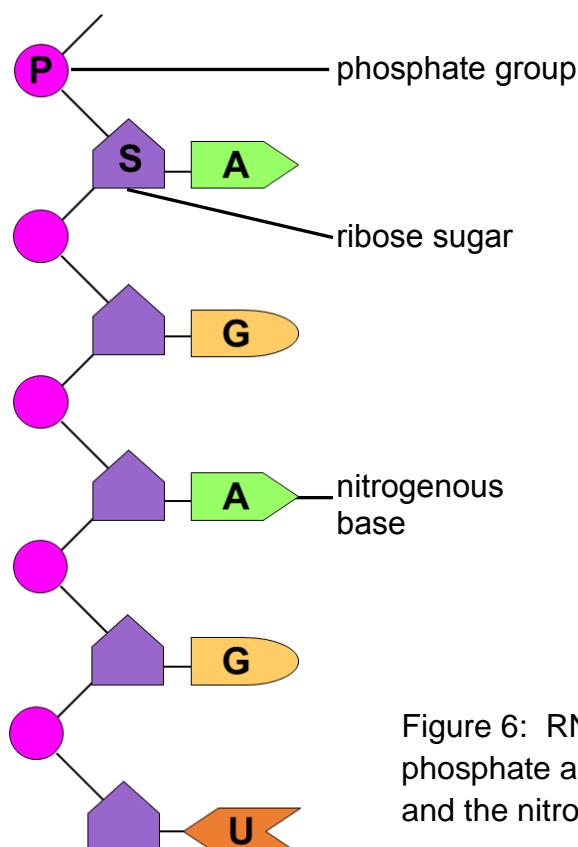


Figure 6: RNA – note the chain formed by the phosphate and sugar molecules on the left, and the nitrogenous bases on the right.

There are four types of nitrogenous bases in RNA:

adenine (A)	cytosine (C)
uracil (U) – <u>not thymine as in DNA</u>	guanine (G)

The role of RNA

The three types of RNA are very important to the process of protein synthesis, with each type playing a unique role.

Comparison between DNA and RNA

DNA and RNA are **similar** in some respects. They both ...

- contain sugar alternating with phosphate
- contain the nitrogenous bases adenine, guanine and cytosine
- play a role in protein synthesis

DNA and RNA also have **significant differences**, tabulated in Table 1 below.

Table 1: The **main differences** between DNA and RNA.

DNA	RNA
contains deoxyribose sugar	contains ribose sugar
double helix and coiled	single stranded
contains the nitrogenous base thymine	contains the nitrogenous base uracil
found in the nucleus only	found in the nucleus, ribosomes and cytoplasm of cells

A comparison DNA and RNA. It is very important to know the differences.

<https://www.youtube.com/watch?v=0Elo-zX1k8M>

DNA replication

DNA replication is the process through which DNA makes an identical copy of itself. This occurs during interphase of the cell cycle in the nucleus. In Figures 7A to 7E, a small portion of DNA is shown undergoing replication.

1. The **DNA double helix** unwinds (Figure 7A)

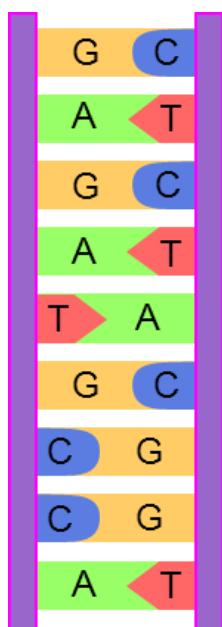


Figure 7A

2. The weak hydrogen bonds between the nitrogenous bases are broken. The DNA strands separate (they **unzip**) (Figure 7B)

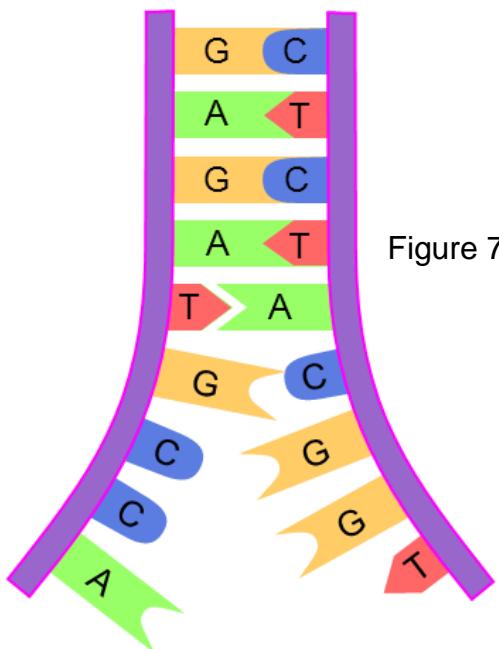


Figure 7B

3. Each original DNA strand serves as a **template** on which its complement is built (Figure 7C)

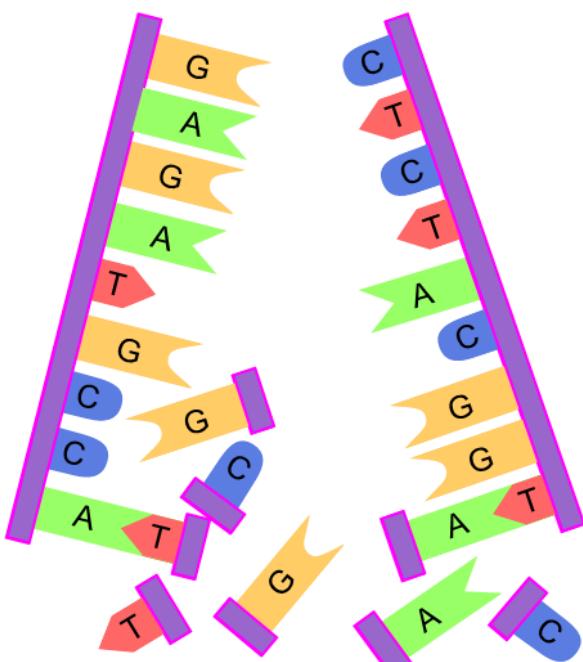


Figure 7C

4. Free nucleotides build a DNA strand onto each of the original DNA strands, attaching their **complementary nitrogenous bases** (A to T and C to G) (Figure 7D)

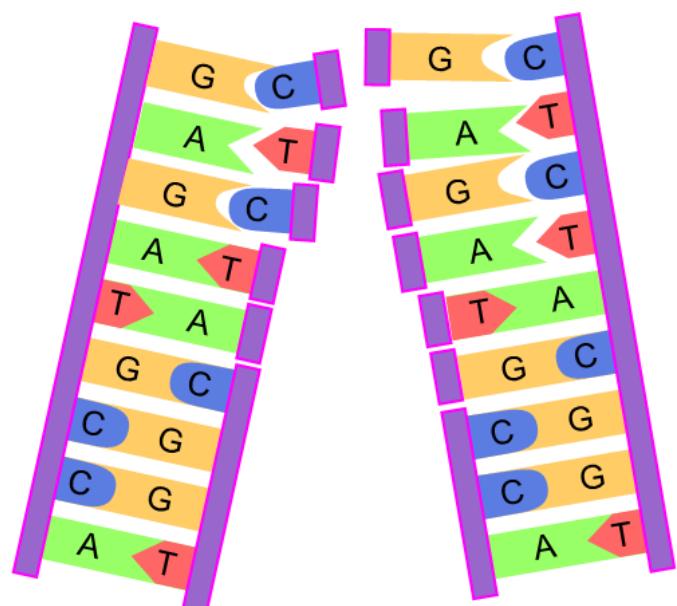


Figure 7D

5. This results in **two identical DNA molecules**. Each molecule consists of one original strand and one new strand (Figure 7E).

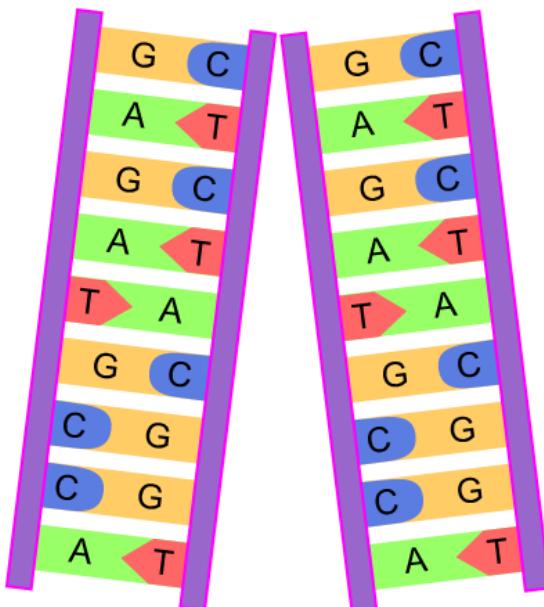


Figure 7E

Figure 7A – 7E: The process of DNA replication

DNA replication is important for cell division, particularly mitosis. It allows each chromosome to be copied so that each new identical daughter cell produced contains the same number and type of chromosomes.

Errors that occur during DNA replication

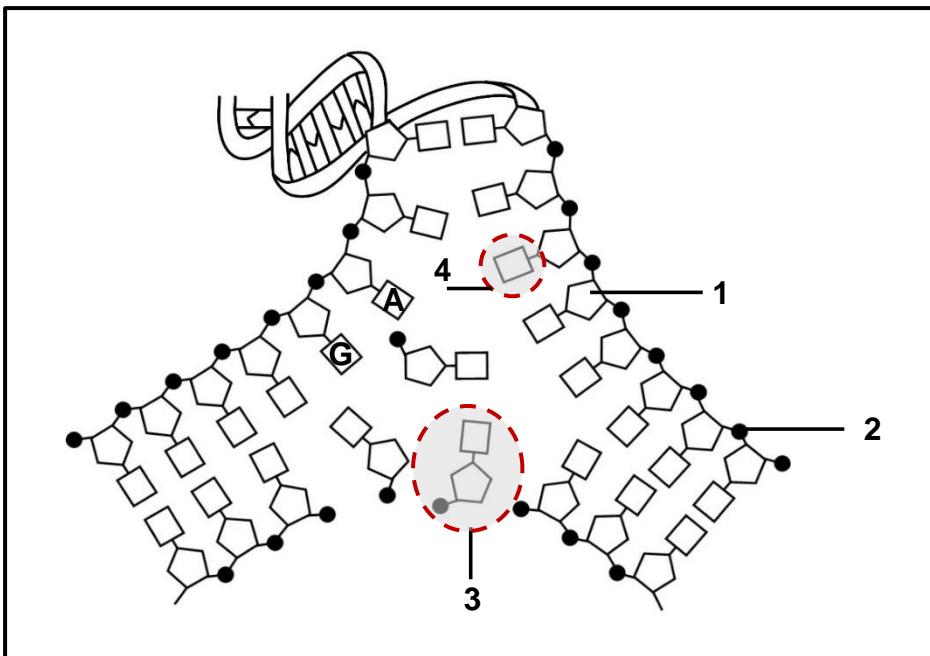
- Errors that occur during DNA replication may sometimes lead to mutations (a change in the nitrogenous base sequence)
- If the incorrect nitrogen base attaches to the original strand and a nitrogen base is added or deleted ...
 - the sequence or order of the bases changes on the new DNA molecule ...
 - resulting in a change in the gene structure

DNA replication: Understand the process.

<https://www.youtube.com/watch?v=Qqe4thU-os8>

Activity 2: DNA replication

Study the diagram below and answer the questions that follow.



1. Name the process illustrated in the diagram above. (1)
2. State the significance of the process mentioned in question 1. (1)
3. Identify the parts labelled as 1, 2, 3 and 4. (4)
4. Describe how this process takes place. (6)
5. Give one location of extra-nuclear DNA. (1)
(13)

DNA profiling

A DNA profile is a pattern produced on X-ray film. This pattern consists of lines which are of different lengths and thicknesses and in different positions, as shown in Figure 8. All individuals, except identical twins, have a unique DNA profile.

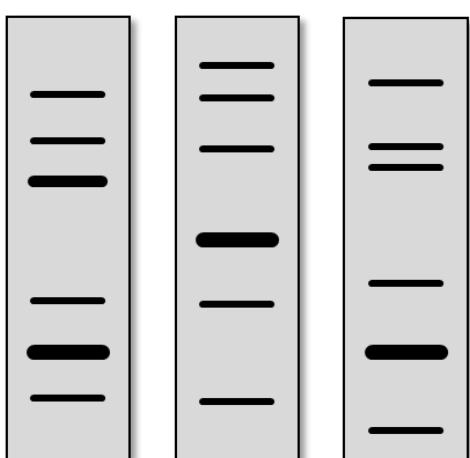


Figure 8: DNA profiles for three different individuals.

DNA profiles are used to:

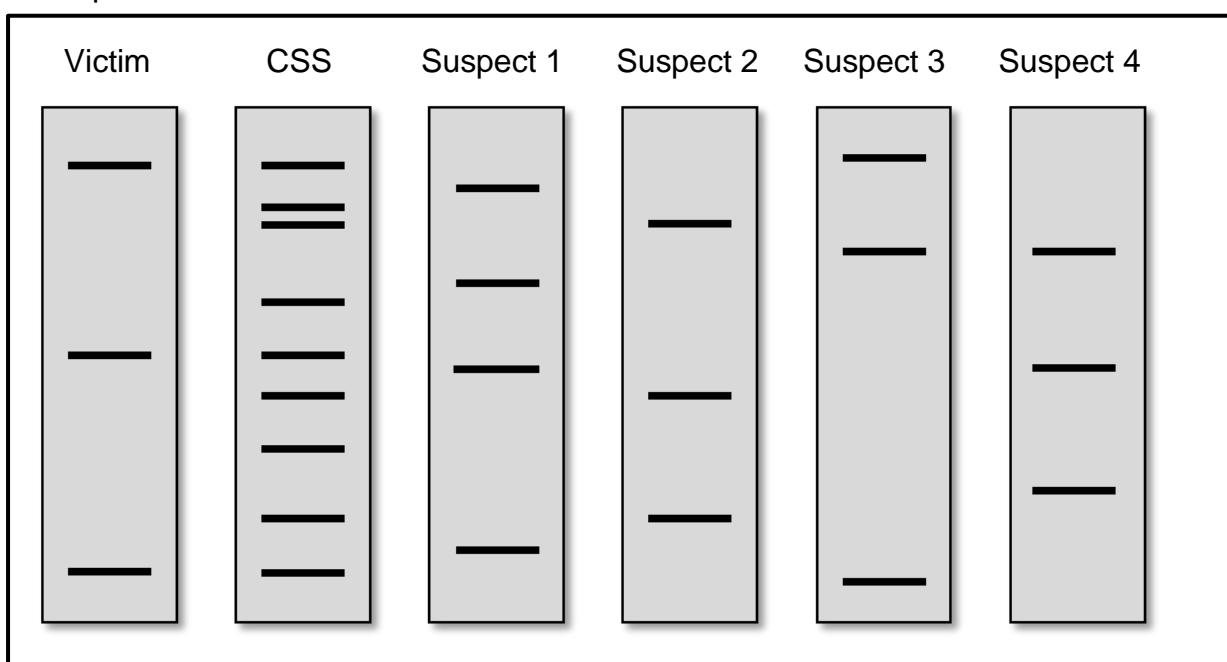
- identify crime suspects in forensic investigations
- prove paternity (father) and maternity (mother) (biological parents)
- determine the probability or causes of genetic defects
- establish the compatibility of tissue types for organ transplants
- identify relatives

DNA profiling is generally accepted as being extremely reliable. The interpretation and comparison of profiles should however be approached with caution, for the following reasons:

- Humans interpret the results which means mistakes could be made
- The method of profiling may be different in different laboratories producing inconsistencies
- Only a small piece of DNA is used in profiling, so the profile might not be 100% unique to a particular individual
- DNA profiling is expensive and therefore not readily accessible to those who cannot afford it, particularly in criminal cases
- DNA profiles may reveal information about a person which could be used against them in a prejudicial way. For example: being HIV positive or having genetic abnormalities may lead to insurance companies not covering a person or prejudice in the court room

Activity 3: DNA profiling

DNA profiles from a crime scene.



In a fight involving a number of people, one person was seriously injured. Police took blood samples from the victim, the crime scene (CSS – crime scene sample) and four suspects. The DNA was then extracted from each sample. The results of these tests are shown in the diagram above.

1. Which suspect probably injured the victim? (1)
 2. Give a reason for your answer to the previous question. (1)
 3. List one application of DNA profiling other than for solving crime. (1)
 4. Give two reasons why DNA profiling may sometimes be challenged. (2)
- (5)

Protein synthesis

Key terminology

amino acids	monomers of proteins
base triplet	three nitrogenous bases one after the other on DNA
transcription	1 st stage of protein synthesis – mRNA formed from DNA carrying code for the protein to be made
translation	2 nd stage of protein synthesis – amino acids combine to form a protein
codon	three nitrogenous bases one after the other on mRNA – these are complementary to the triplet on DNA
anti-codon	three nitrogenous bases one after the other on tRNA – these are complementary to the codon on mRNA

The process in which proteins are made is called protein synthesis. Proteins are made by linking various amino acids that are present in the cytoplasm of cells. There are 20 different amino acids, and they combine in a large variety of combinations. The number of amino acids and the sequence of the amino acids determine the type of protein that is formed.

Figure 9 illustrates a protein with different amino acids represented by the different shapes and colour. The bond between the amino acids is known as a peptide bond.

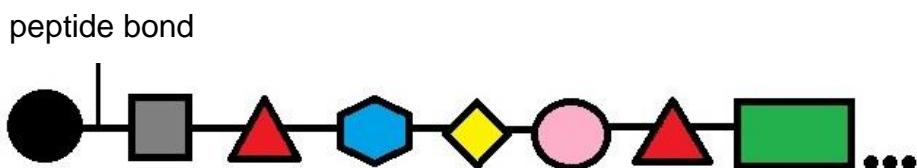


Figure 9: Amino acids linked by peptide bonds

The genes found in DNA contain the code which determines which type of protein that will be formed.

- The smallest protein contains 50 amino acids linked together
- Proteins generally contain 300 or more amino acids.

Three consecutive nitrogenous bases on the DNA strand are called the base triplet. The base triplets determine which amino acid will be placed into the protein as well as the sequence in which the amino acids will be joined.

Protein synthesis occurs in two stages

Stage 1: Transcription

Stage 2: Translation

Stage 1: Transcription

The first stage of protein synthesis, called transcription, occurs in the nucleus (see Figure 10 below).

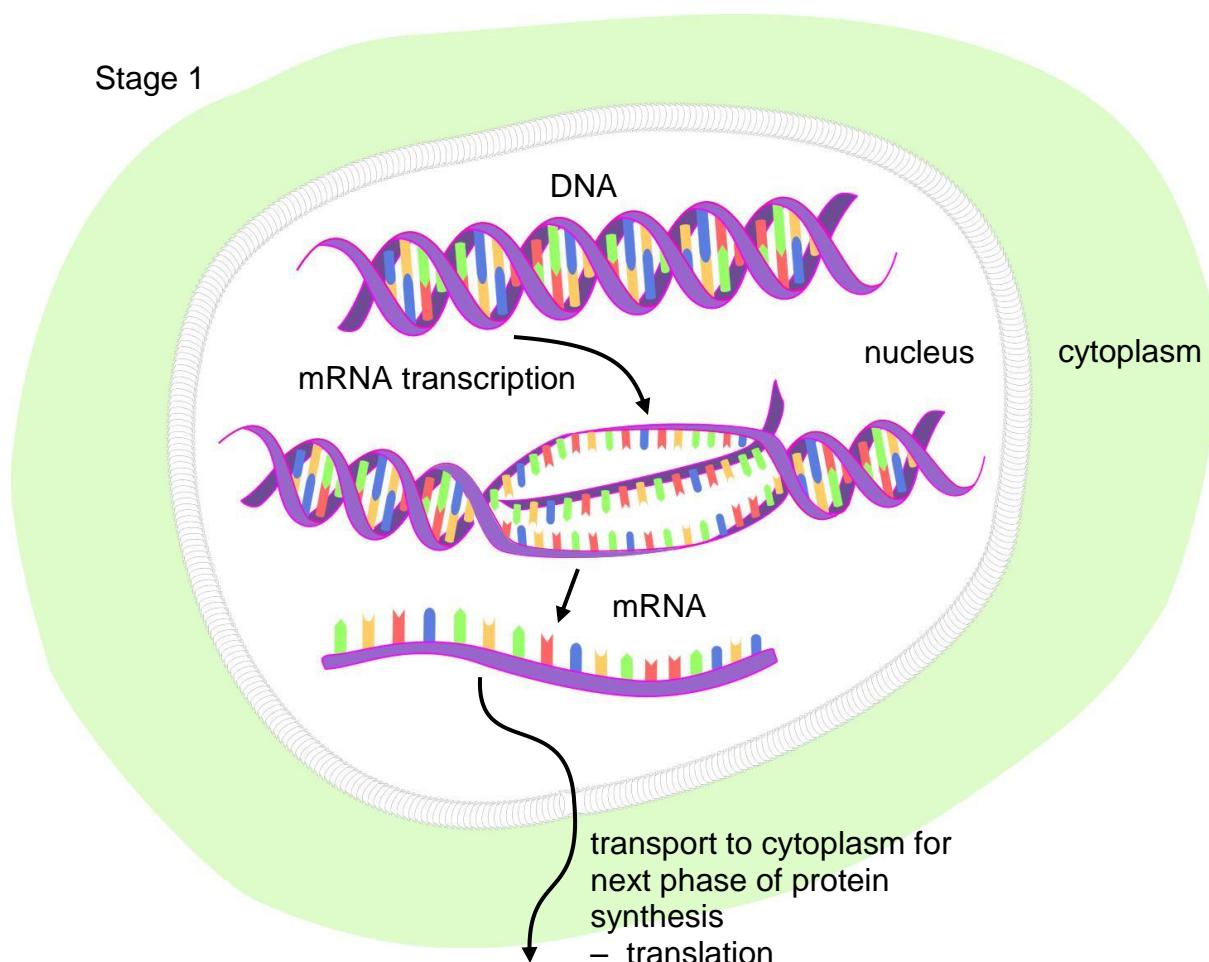
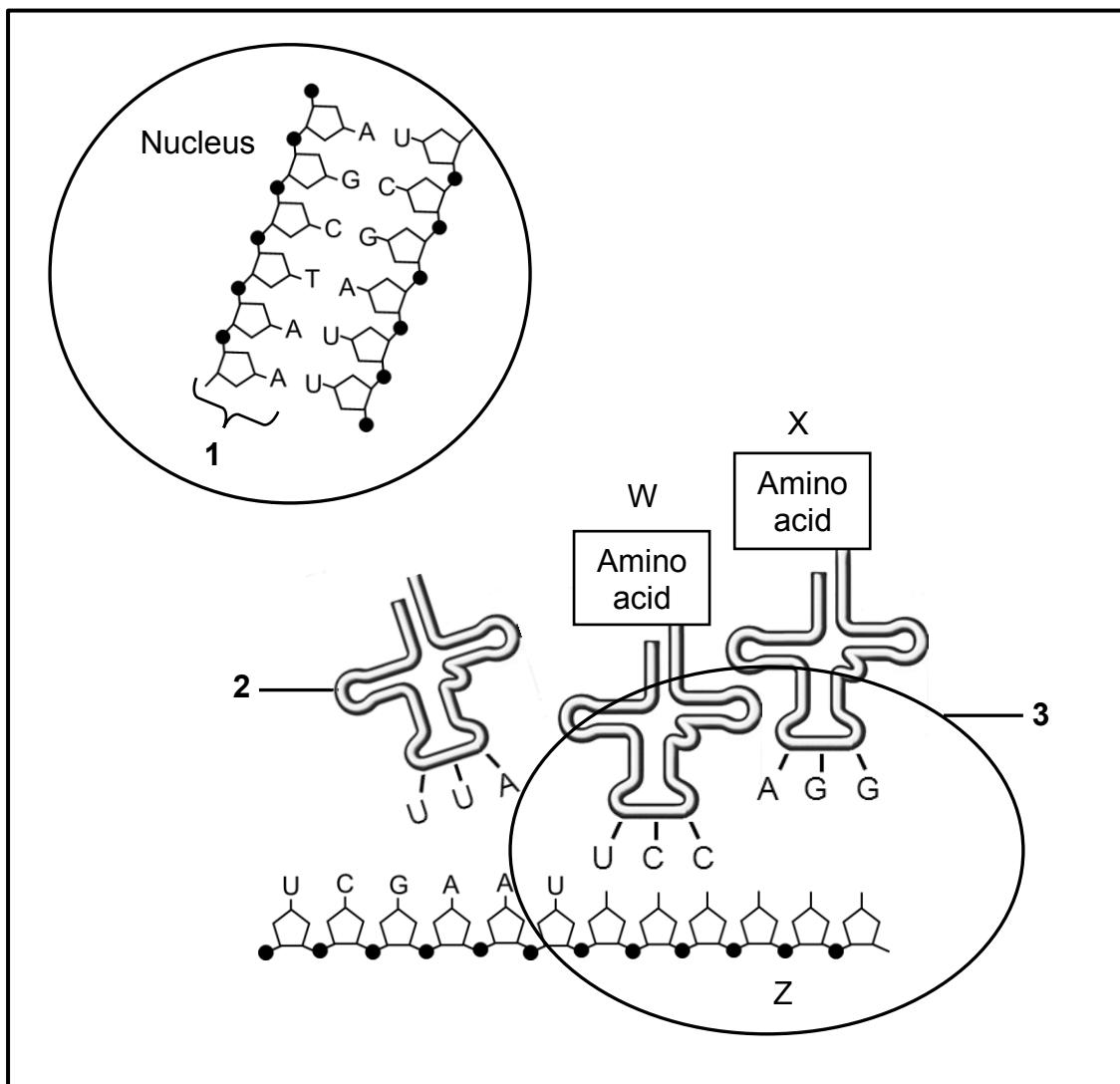


Figure 10: Transcription

- 2.1.3. Name and describe the process occurring in the nucleus which results in the formation of an mRNA molecule. (6)
- 2.1.4. Draw a RNA nucleotide with a complementary base to adenine. (2)
- (14)

2.2 The diagrams below represent the process of protein synthesis. Study them and answer the questions that follow.



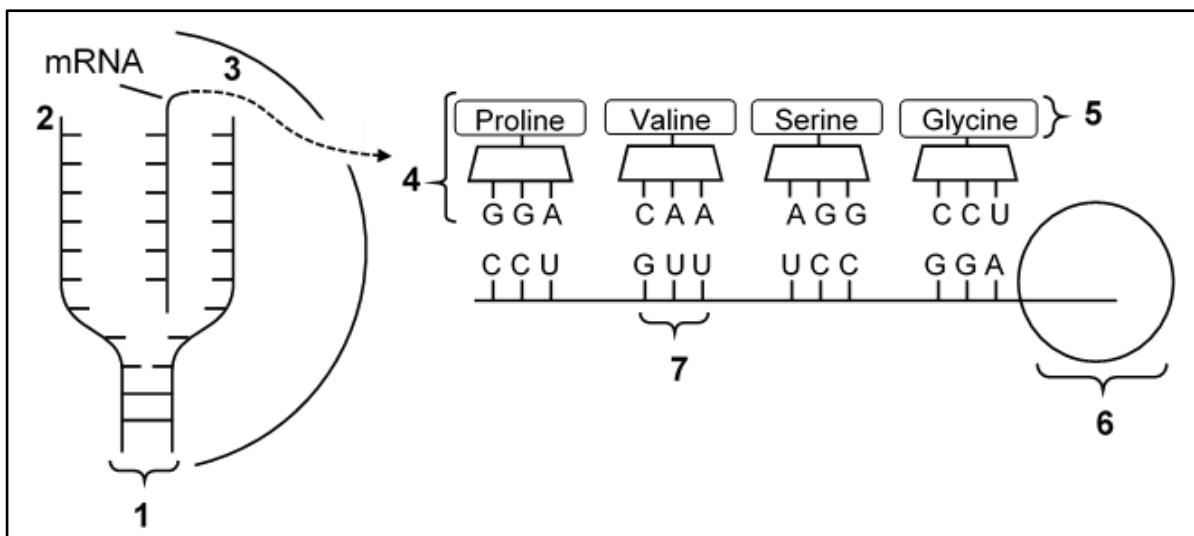
- 2.2.1 Identify the structures labelled **1,2** and **3**. (3)
- 2.2.2 Name and describe the stage of protein synthesis taking place at **Z** (5)
- 2.2.3 Using the table below, work out the names of the amino acids labelled **W** and **X**. (4)

Base Triplet on mRNA coding for the amino acid	Amino acid coded for
GAG	glutamate
CAG	histidine
AGG	arginine
CUG	leucine
UCC	proline
GUG	valine

(12)
[26]

Question 3

3.1 The diagram below represents two stages of protein synthesis.



3.1.1 Provide labels for:

- a) molecule 1 (1)
- b) organelle 6 (1)

3.1.2 Give only the number of the part which represents a:

- a) DNA template strand (1)
- b) monomer of proteins (1)
- c) codon (1)

3.1.3 Describe *translation* as it occurs in organelle 6. (4)

3.1.4 Provide the:

- a) DNA sequence that codes for glycine (2)

- b) codon for proline (2)
- 3.1.5 State two differences between a **DNA** nucleotide and an **RNA** nucleotide. (4)
- (17)

- 3.2 The first 7 triplets of nitrogenous bases that form part of the gene coding for one chain of the haemoglobin protein that makes up red blood corpuscles in humans is shown below. Study the table and answer the questions that follow.

DNA Template	CAC	GTG	GAC	TGA	GGA	CTC	CTC
Base triplet number	1	2	3	4	5	6	7

- 3.2.1 How many of the following are coded for in the DNA template sequence above?
- a) Nitrogenous bases (1)
 - b) Different types of tRNA molecules that are required to form the polypeptide from this piece of DNA. (1)
- 3.2.2 Write down the mRNA sequence for the triplets numbered **4** and **6** in the above table. (2)
- 3.2.3 Using the table below, determine the amino acid sequence coded for by triplet numbers **4** and **6**. (2)

Anticodons on tRNA coding for the amino acid	Amino acid coded for
CUC	glutamate
GUC	histidine
GGA	proline
GAC	leucine
UGA	threonine
CAC	valine

- 3.2.4. If the T in the 6th base triplet changed to A in the DNA template above, write down the new amino acid (using the table above) that this 6th triplet now codes for. (1)
(7)
[24]

Section B: [50]

Total Marks: [100]

2: Meiosis

Introduction	Prophase II
Significance of DNA replication for meiosis	Metaphase II
The karyotype	Anaphase II
Meiosis – the process	Telophase II
Introduction	Comparing mitosis and meiosis
The process of meiosis	Activity 1: Meiosis I and Meiosis II
First meiotic division	Abnormal meiosis (chromosome mutation)
Prophase I	Chromosome mutations
Metaphase I	Enrichment
Anaphase I	
Telophase I	
Second meiotic division	End of topic exercises

CHAPTER 2: MEIOSIS

Introduction

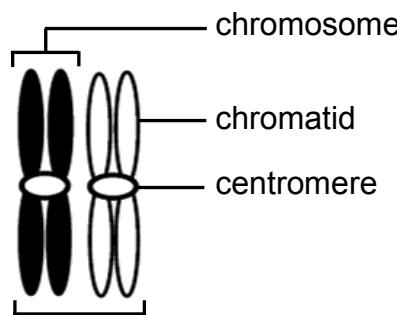
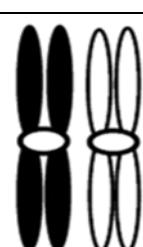
In Grade 10 you learnt that when a cell divides by **mitosis**, two exact copies of the mother cell are produced. Cells divide by mitosis for the purpose of growth and repair. Worn out or damaged cells are replaced by the mitotic division of somatic cells (body cells) and some organisms are able to reproduce asexually by mitosis.

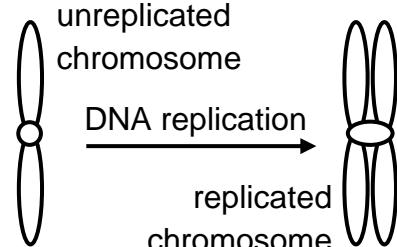
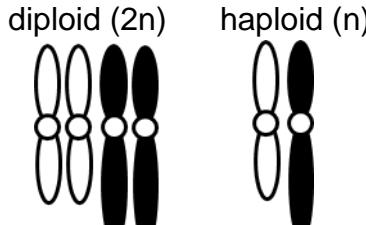
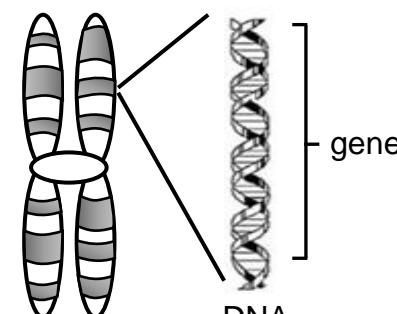
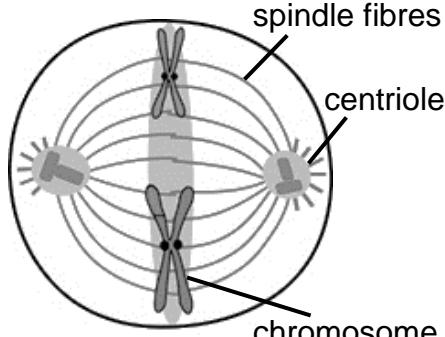
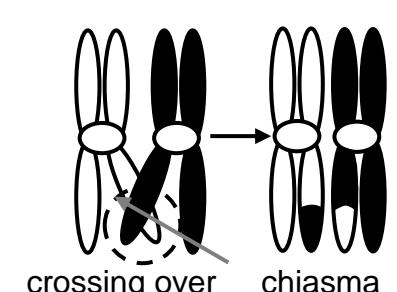
Meiosis is a special type of cell division that halves the number of chromosomes. Four genetically different haploid daughter cells are formed from one diploid cell.

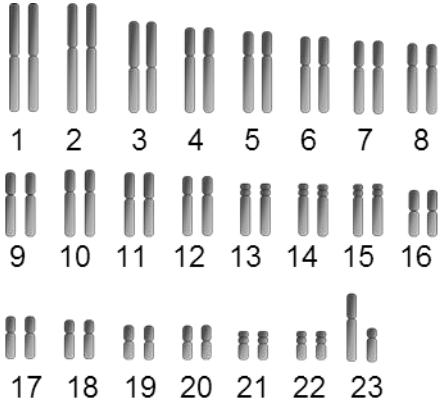
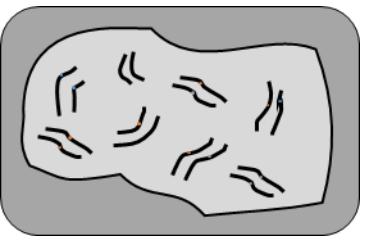
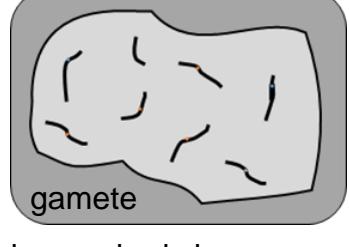
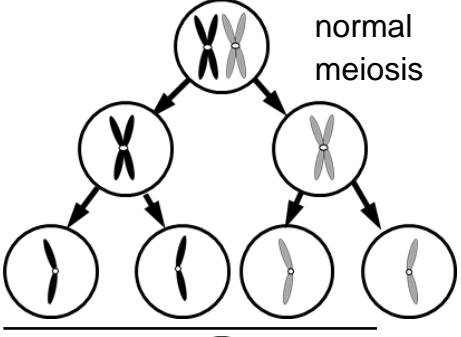
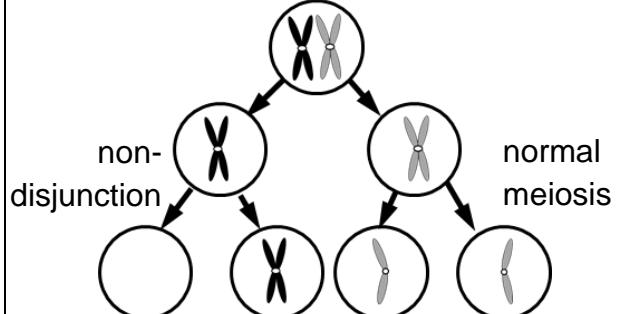
Meiosis is important because

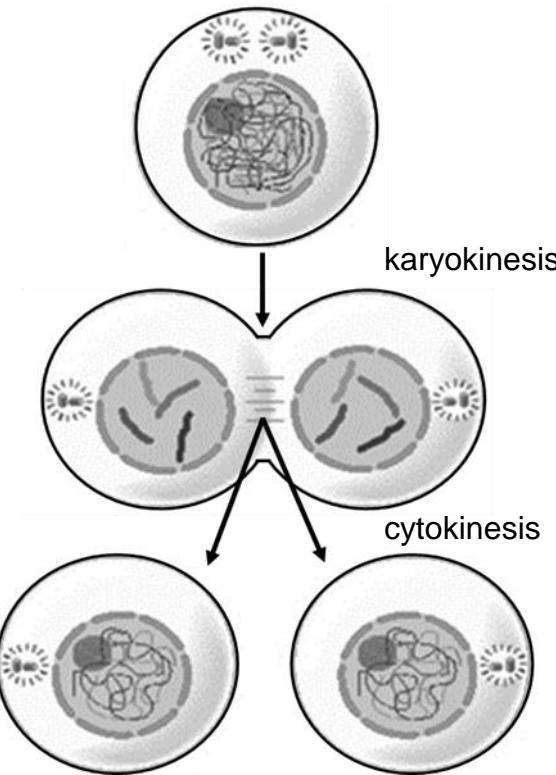
- haploid gametes are produced
- the doubling effect of fertilization on chromosome number of future generations is overcome
- genetic variation occurs

Key terminology

chromosome	a threadlike structure made up of DNA and protein found in the nucleus of most living cells, carrying genetic information in the form of genes	
chromatid	one of the two identical strands of a replicated chromosome	
centromere	region where the two chromatids of a chromosome are held together	
homologous chromosomes	a pair of chromosomes of the same shape, size and having similar genes for each characteristic occupying the same position	homologous chromosomes – one from the mother and one from the father
bivalent	a pair of homologous chromosomes which lie next to each other and are physically in contact with each other at a point where crossing over will occur	

unreplicated chromosomes	an unreplicated "chromosome" has a single double-stranded DNA molecule	
replicated chromosomes	a replicated "chromosome" has two identical double-stranded DNA molecules	
interphase	the phase in the cell cycle when DNA replication occurs	
diploid (2n)	two complete set of chromosomes in a cell	
haploid (n)	one complete sets of chromosomes in a cell	
gene	a segment of DNA in a chromosome that contains the code for a particular characteristic	
centrosome	organelle (containing two centrioles) found only in animal cells	
centriole	structures formed when the centrosome divides into two; they move to opposite ends of the cell during cell division	
crossing over	Overlapping of homologous chromosomes resulting in the exchange of genetic material during Prophase I	
chiasma	point where two chromatids overlap during crossing over	

karyotype	a representation of the number, shape and arrangement of a full set of chromosomes in the nucleus of a somatic cell	
autosome	the first 22 pairs of chromosomes which control the appearance, structure and functioning of the body	
gonosomes (sex chromosomes)	the pair of chromosomes (XX or XY) responsible for sex determination	
somatic cells (body cells)	Any cells in an organism excluding male and female gametes – they are diploid (have 2 sets of chromosomes) and are produced through mitosis	 somatic cell – chromosomes are in homologous pairs
sex cells (gametes)	specialized cells called gametes (sperm cell and egg cell). They have a haploid number of chromosomes and are produced through meiosis	 single unpaired chromosomes
non-disjunction	when homologous chromosome pairs fail to separate in meiosis	 

karyokinesis	Karyo means “nucleus” and kinesis means “synthesis or division.” Karyokinesis is the process of division of the nucleus of a cell	
cytokinesis	Cyto means “cytoplasm,” and kinesis mean “synthesis or division.” Cytokinesis is the process of division during which the cytoplasm of a single cell divides into two daughter cells.	

Significance of DNA replication for meiosis

Each species of living organism has a characteristic number of chromosomes found floating in the nucleoplasm of the nucleus.

- When the cell is not dividing, the genetic material forms a tangled chromatin network.
- During interphase, DNA replication takes place.
- Single-stranded chromosomes become double-stranded.
- Each chromosome will now consist of two chromatids joined by a centromere.
- This ensures the sharing of the hereditary material by all the daughter cells that will be formed.

Meiosis – the process

Introduction

If meiosis does not occur, gametes will not be formed and sexual reproduction would not be able to take place.

- During meiosis haploid gametes are formed, these gametes have half the number of chromosomes.
- This is important so that the chromosome number is **not doubled** when fertilisation occurs. Instead, when two haploid gametes fuse, the **diploid** number is restored.
- Meiosis thus ensures that the same number of chromosomes within a species remains constant from generation to generation.

In animals, meiosis occurs in the sex organs (ovaries and testes) to produce gametes (gametogenesis).

In plants meiosis produces spores which are used for reproduction in mosses and ferns.

In flowering plants (angiosperms) meiosis takes place in the anther and in the ovule.

The process of meiosis

NOTE that a human cell has 46 chromosomes arranged in 23 pairs of homologous chromosomes. Explaining meiosis using a human cell is very complex. The same principle, however, applies to all somatic cells so for the sake of simplicity, meiosis will be explained using a cell with only 4 chromosomes.

In meiosis, the cell undergoes two divisions which are referred to as **Meiosis I** and **Meiosis II**.

- **Meiosis I** is a reduction division which reduces the diploid number of chromosomes to haploid.

First meiotic division

Although meiosis is a continuous process, the events are placed into phases for convenience.

Prophase I

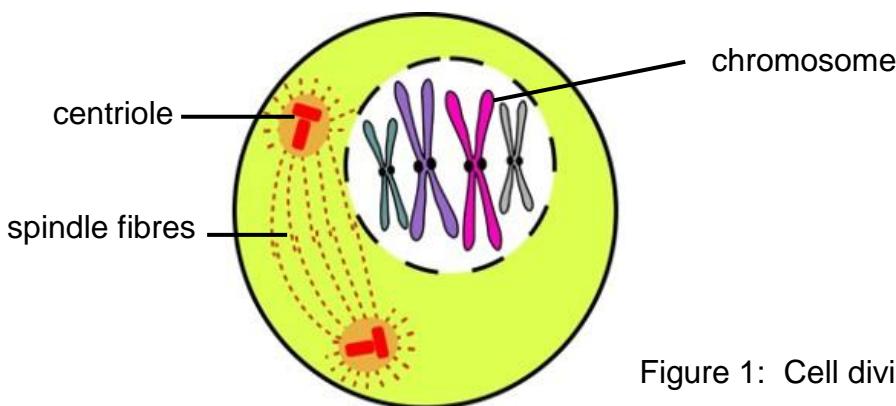


Figure 1: Cell dividing in Prophase I

- Nuclear membrane and nucleolus start to disappear.
- Centrosome splits and the two centrioles move apart forming spindle fibres.
- Chromatin network condenses into individual chromosomes and pairs of homologous chromosomes lie next to each other forming a bivalent.
- Inner chromatids from each homologous chromosomes overlap and touch each other at a point called the chiasma (plural: chiasmata) in a process called crossing over (see Figure 2)
- Chromatid segments break off and are exchanged, resulting in the exchange of genetic material.
- This process is called crossing over and it brings about variation.

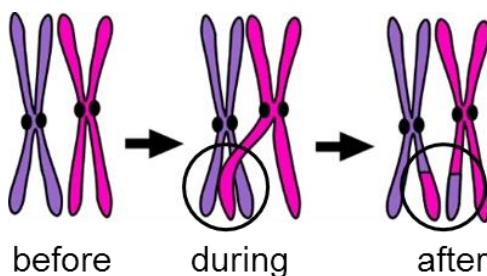


Figure 2: The process of crossing over

Metaphase I

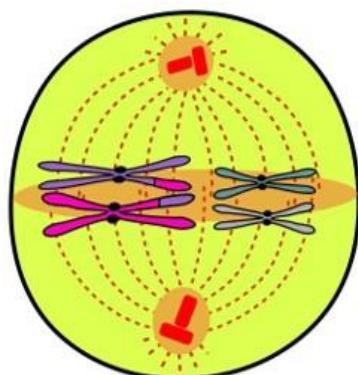


Figure 3: Cell undergoing Metaphase I

- Homologous chromosomes move to the middle of the cell (the equator). The two homologous chromosomes lie on opposite sides of the equator parallel to each other (Figure 3).
- Which chromosome lies on which side of the equator is totally up to chance. This is called **random arrangement** and brings about further variation.
- Each chromosome in the pair becomes attached to a spindle thread by the centromere.

Anaphase I

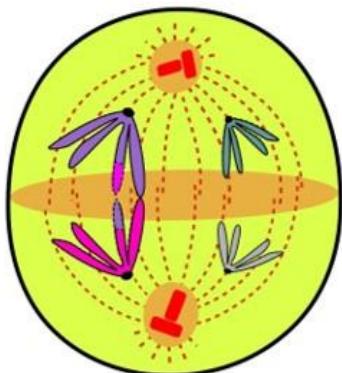


Figure 4: Cell undergoing Anaphase I

- One **whole chromosome** from each pair is pulled to opposite poles by contraction of the spindle fibres (see Figure 4).
- This separates the homologous chromosomes – one to each pole.

Telophase I

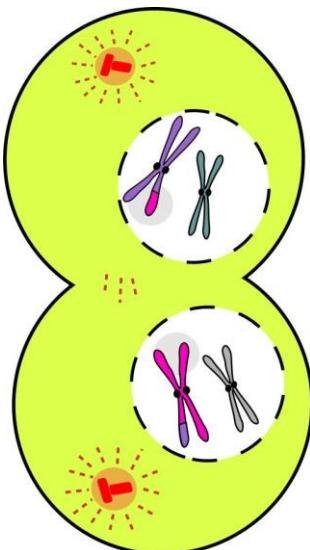


Figure 5: Cell undergoing Telophase I

- A new nuclear membrane forms around the group of chromosomes at each pole (Figure 5).
- Nucleolus returns.
- Cytokinesis (division of cytoplasm) splits the mother cell into two daughter cells.

Important: Each daughter cell now has **half** the number of chromosomes and each has a slightly different genetic make-up due to crossing over.

Second meiotic division

The second meiotic division takes place in **both** daughter cells formed during Meiosis I.

Prophase II

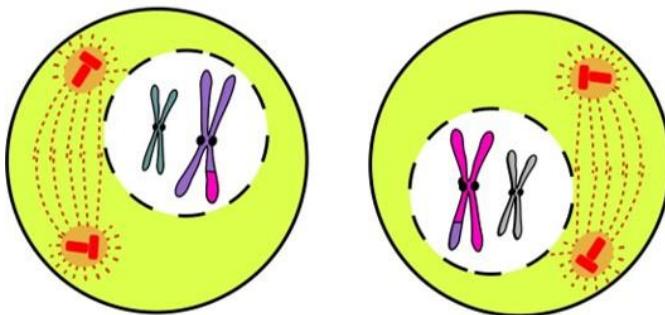


Figure 6: Cells undergoing Prophase II

- Nuclear membrane and nucleolus start to disappear.
- Centrosome splits into two centrioles and a spindle forms.
- Chromosomes are NOT in pairs (Figure 6).

Remember: Each chromosome is made of TWO chromatids.

Metaphase II

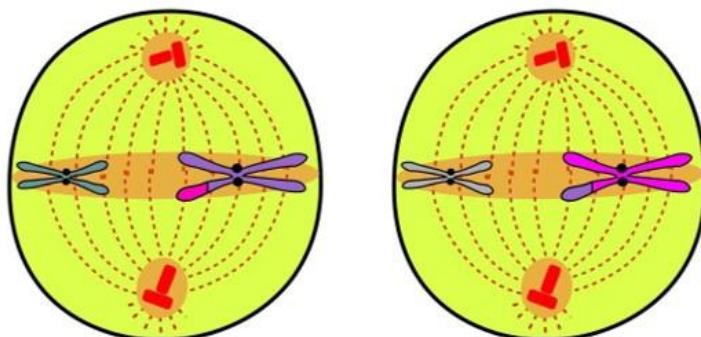


Figure 7: Cells undergoing Metaphase II

- Single chromosomes arrange themselves randomly along the equator with the centromere in line with the equatorial plane (Figure 7). Which chromatid faces which pole is totally up to chance.
- Each chromosome becomes attached to a spindle fibre.

Anaphase II

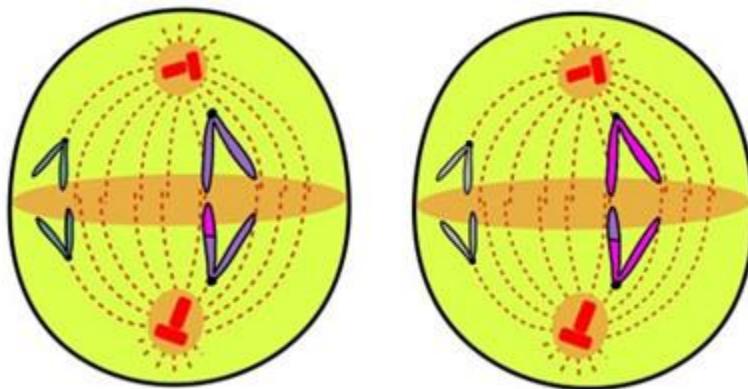


Figure 8: Cell undergoing Anaphase II

- The centromere splits and the two chromatids are pulled to opposite poles (Figure 8).

Telophase II

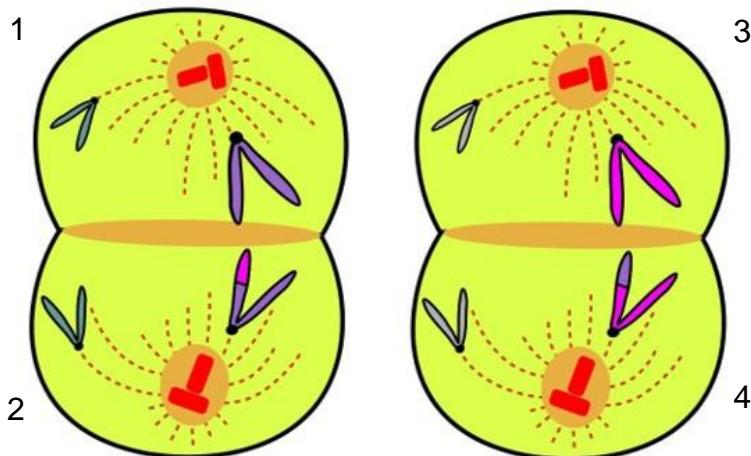


Figure 9: Cells at start of Telophase II

- A new nuclear membrane forms around the unreplicated chromosomes at each pole (Figure 9).
- Cytokinesis splits the cell into two new cells (Figure 10).

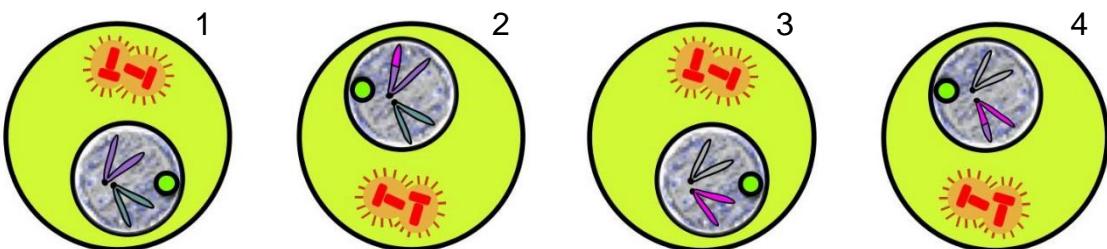


Figure 10: Daughter cells at the end of Telophase II

Important: As Meiosis II took place in TWO cells, there will now be FOUR daughter cells. These cells will be haploid and genetically different to each other.

The four stages of meiosis can be remembered in the following way - **PMAT**

Prophase – chromosome **PAIR** up (crossing over).

Metaphase – chromosomes move to **MIDDLE**.

Anaphase – chromosomes move **APART** to the poles.

Telophase – **TERMINAL** phase where daughter cells are formed.

Why is ‘Crossing over’ important?

- Crossing over brings about an exchange of genetic material during the process of gamete formation which results in the formation of new genetic combinations.
- This results in formation of gametes that will give rise to individuals that are **genetically different** from their parents and siblings.

Meiosis made SUPER EASY:

https://www.google.co.za/search?q=videos+on+meiosis&rlz=1C1AZAA_enZA747ZA747&oq=videos+on+meiosis&aqs=chrome..69i57j0l4.6731j0j8&sourceid=chrome&ie=UTF-8

Importance of meiosis

- Production of gametes (four daughter cells are formed).
- Halving of the chromosome number (diploid to haploid) so that the chromosome number remains constant from generation to generation within a species.
- Mechanism to introduce genetic variation through:
 - Crossing over during Prophase I.
 - The random arrangement of chromosomes at the equator during Metaphase I and II as can be seen in Figure 11 below.

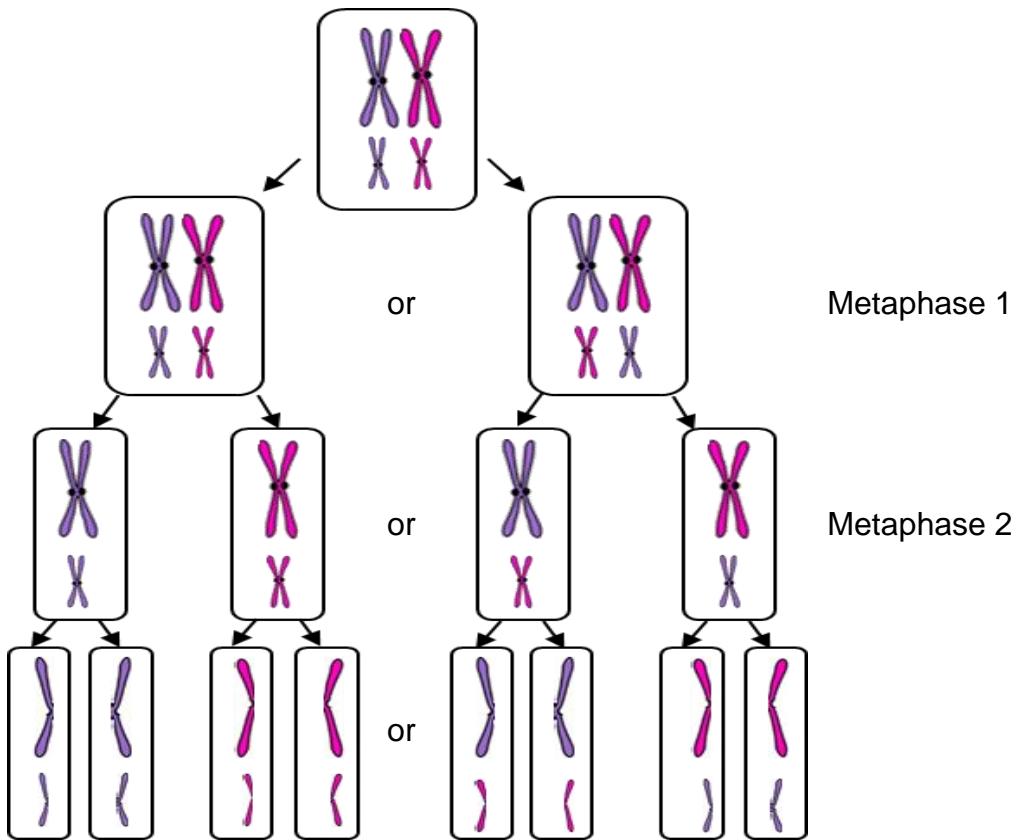


Figure 11: Random arrangement of chromosomes

Differences between meiosis I and meiosis II

Table 1: The differences between Meiosis I and Meiosis II

Meiosis I	Meiosis II
Chromosomes arrange at the equator of the cell in homologous pairs	Chromosomes line up at the equator of the cell individually
Whole chromosomes move to opposite poles of the cell	Chromatids move to opposite poles of the cell
Two cells are formed at the end of this division	Four cells are formed at the end of this division
The chromosome number is halved during meiosis I (diploid → haploid)	The chromosome number remains the same (haploid) during meiosis II
Crossing over takes place	Crossing over does not take place

Comparing mitosis and meiosis

Table 2: Showing the differences between mitosis and meiosis

Mitosis	Meiosis
Mitosis occurs in body cells	Meiosis occurs in sex organs
Both karyokinesis and cytokinesis occurs once	Both karyokinesis and cytokinesis occurs twice
Two daughter cells are formed	Four daughter cells are formed
Daughter cells are genetically identical to one another and to the parent cell	Daughter cells are genetically different from each other and from the parent cell
Chromosome number remains constant	Chromosome number is halved
Crossing over does not occur	Crossing over occurs

The differences between mitosis and meiosis as a rap song:

<https://youtu.be/qH4WUUQ5pOI>

- The only similarities are between mitosis (Prophase, Metaphase and Anaphase) and meiosis II (Prophase II, Metaphase II and Anaphase II).

The differences between mitosis and meiosis are illustrated in the following diagram.

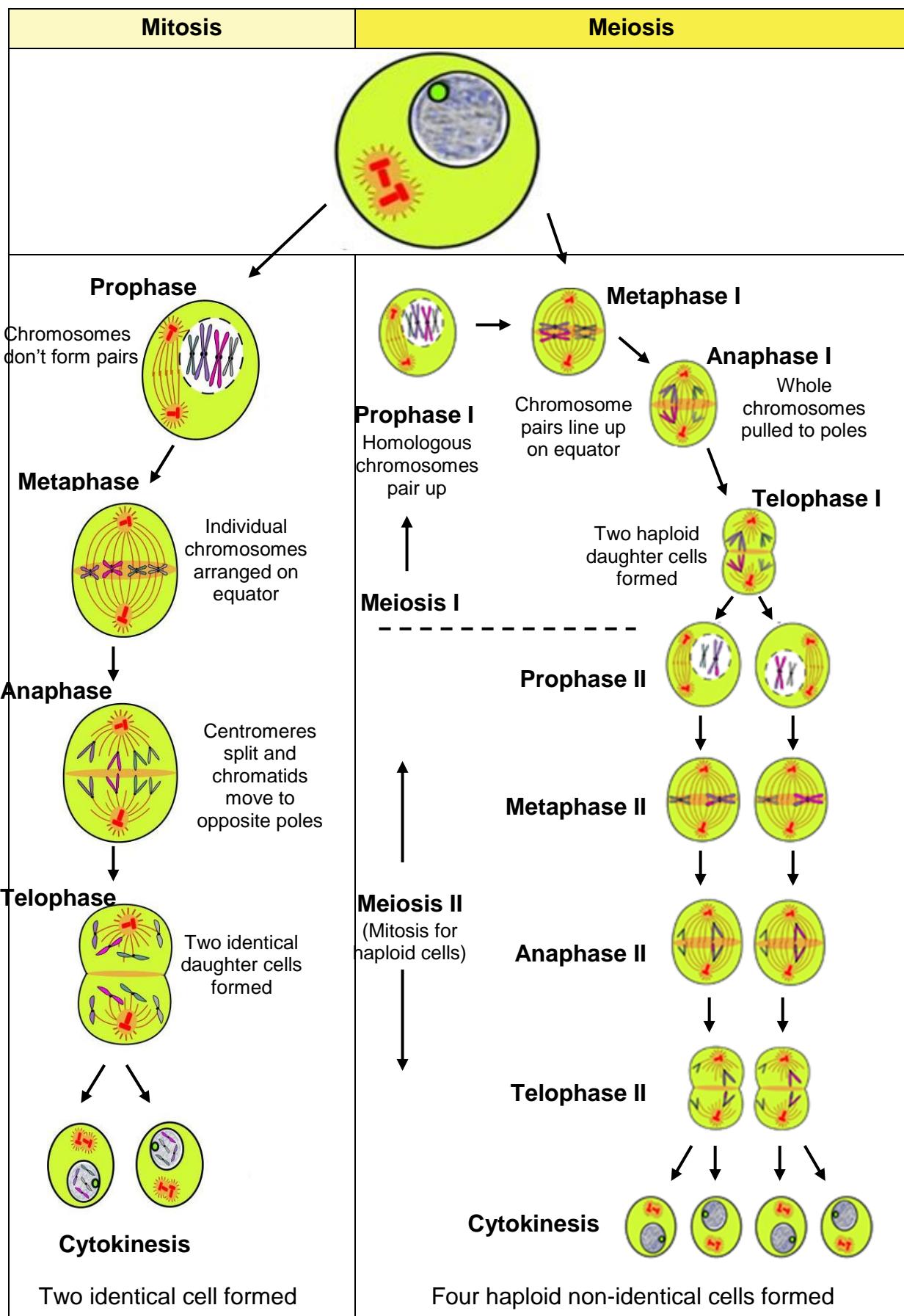


Figure 12: The difference between MITOSIS and MEIOSIS

Activity 1: Meiosis I and Meiosis II

1. Various options are provided as possible answers to the following questions. Choose the correct answer and write only the letter (A – D) next to the question number (1.1.1 – 1.1.5) on your answer sheet, for example 1.1.6 D

- 1.1 Which one of the following correctly describes the daughter cells produced by meiosis?

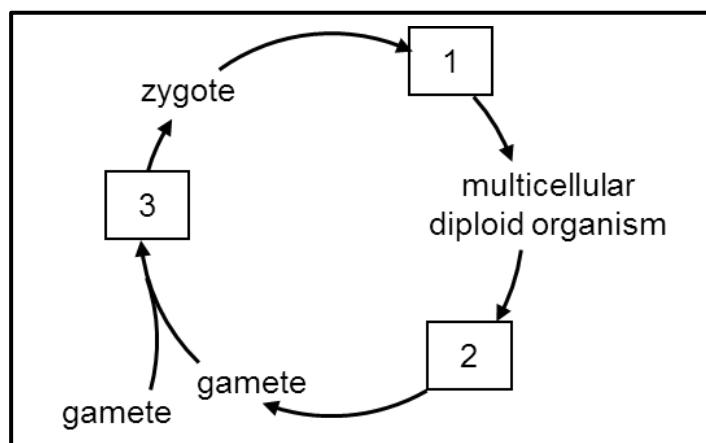
Cells produced by meiosis

	Chromosome number	Genetic composition
A	haploid	different
B	diploid	identical
C	diploid	different
D	haploid	identical

- 1.2 If there are 38 chromosomes in the body cell of a donkey. How many of these chromosomes are autosomes?

A 38 B 19 C 36 D 44

- 1.3 Use the sketch below to identify processes 1, 2 and 3.

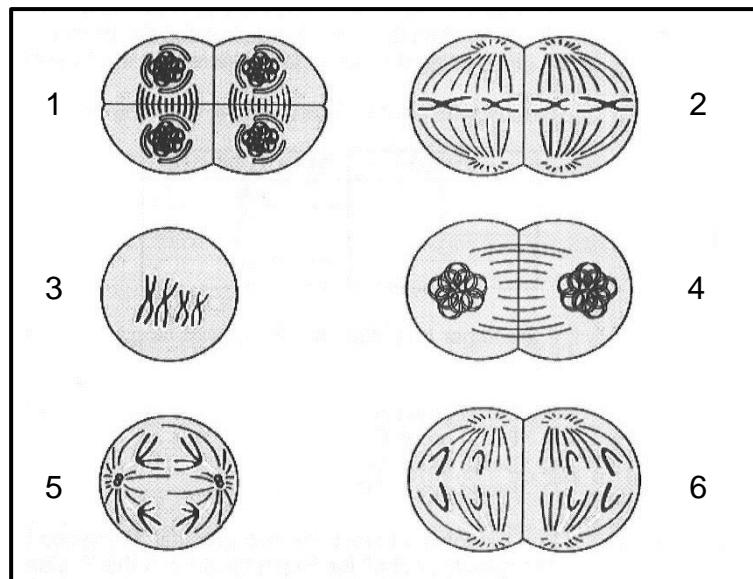


	1	2	3
A	meiosis	fertilisation	mitosis
B	fertilisation	mitosis	meiosis
C	mitosis	meiosis	fertilisation
D	fertilisation	meiosis	mitosis

- 1.4 Cytokinesis is a term that describes ...

A nuclear division
B cytoplasmic division
C reduction of the chromosome number
D doubling the chromosome number

- 1.5 The diagrams below represent six different phases of meiosis taking place in a particular cell.

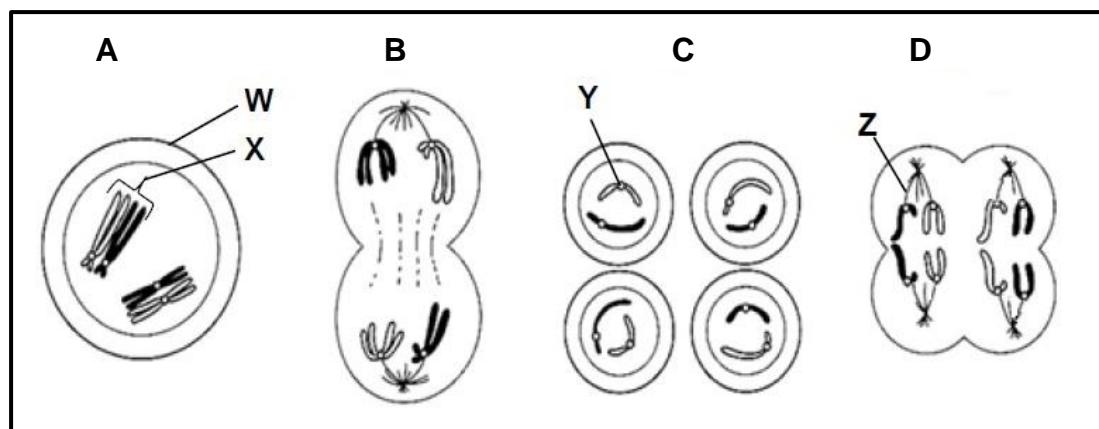


- 1.5.1 The diploid number of chromosomes in this cell is ...
 A 2 B 4 C 8 D 46
- 1.5.2 The correct sequence from the start of meiosis till the end is ...
 A 1, 2, 3, 4, 5, 6 B 6, 2, 5, 4, 1, 3
 C 3, 5, 4, 2, 6, 1 D 3, 4, 5, 6, 1, 2
- 1.6 Interphase is the stage during which ...
 A nothing happens in the cell.
 B a dividing cell forms a spindle.
 C cytokinesis occurs.
 D a cell grows and duplicates its DNA. $(7 \times 2) = (14)$

2. Each of the following questions consist of a statement in Column I and two items in Column II. Decide which item(s) relate(s) to the statement. Write **A only**, **B only**, **Both A and B** or **None** next to the question number.

	Column I	Column II
2.1	Chromosome number changes from diploid to haploid	A: Meiosis B: Mitosis
2.2	Takes place to form sex cells	A: Mitosis B: Meiosis
2.3	Replication of DNA takes place	A: before mitosis B: before meiosis

- 1.5 The diagrams below show different phases in meiosis. Study the diagrams and answer the questions that follow.



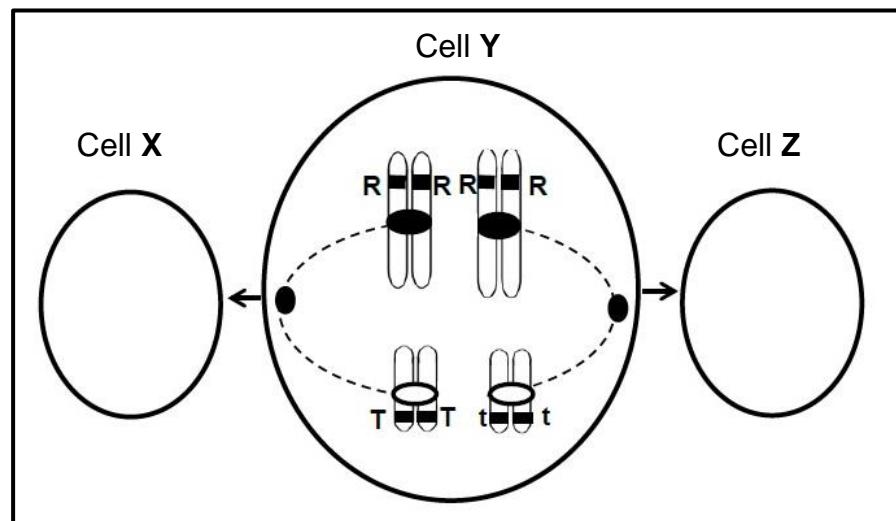
- 1.5.1 Label structures **W** and **X**. (2)
- 1.5.2 How many chromosomes are present in each cell:
- phase **A** (1)
 - phase **C** (1)
- 1.5.3 Give the letter of the diagram that represents Anaphase II. (1)
- 1.5.4 State the name and function of region **Y** and structure **Z**. (4)
- 1.5.5 Which phase precedes (occurs before) phase **A**? (1)
- (10)

Section A: [50]

Section B

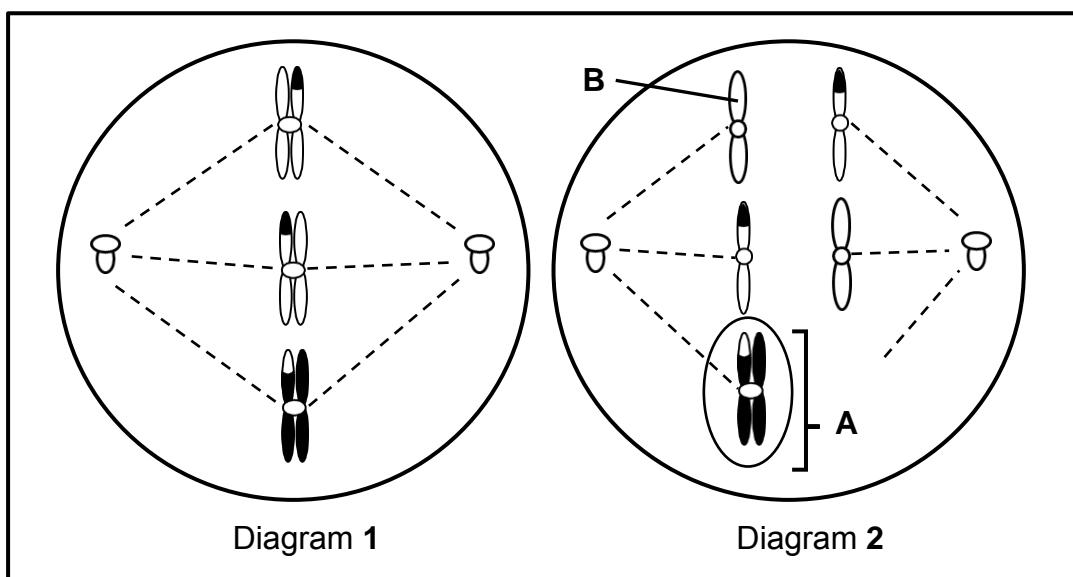
Question 2

- 2.1 The diagram below represents a phase in meiosis. Cell **Y** undergoes division to give rise to cells **X** and **Z**. Some alleles are indicated by letters.



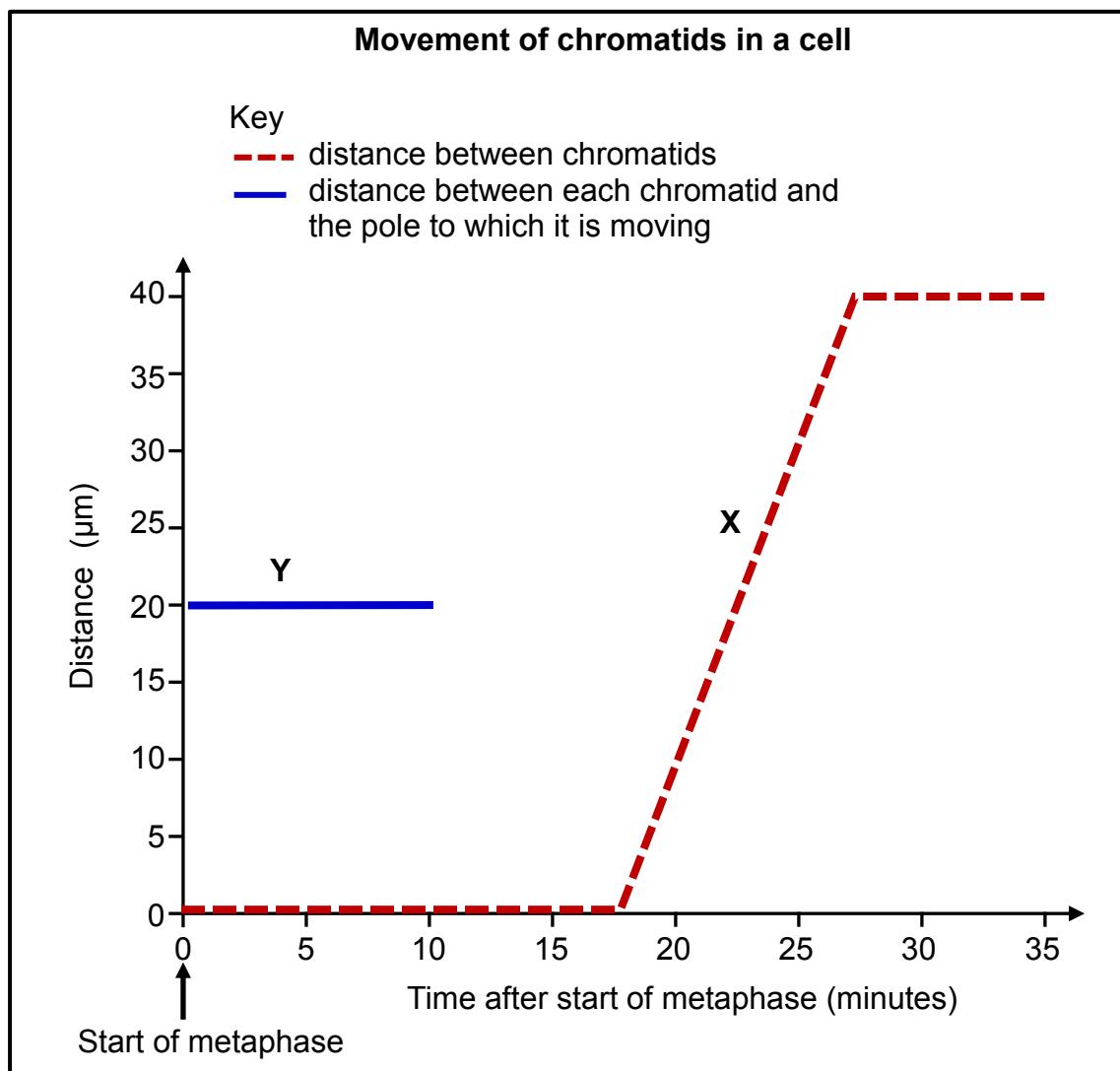
- 2.1.1 Explain why cell **Y** does not belong to a human. (2)
- 2.1.2 How many chromosomes would be present in:
- cell **X** at the end of Telophase I. (1)
 - the daughter cells produced by cell **Z** after meiosis II. (1)
- 2.1.3 Draw a labelled diagram of a gamete that will result from cell **Y**. (5)
- 2.1.4 Describe the events of Anaphase II. (3)
- (12)

- 2.2 Study the diagrams below representing two phases of meiosis and answer the questions that follow.



- 2.2.1 Identify the phase represented by:
- Diagram 1 (1)
 - Diagram 2 (1)
- 2.2.2 Name the part labelled **B**. (1)
- 2.2.3 Describe what happens during the phase illustrated in Diagram 1. (2)
- 2.2.4 In Diagram 2 the part circled, and labelled **A** is an abnormality during the process of meiosis.
- Name this abnormality. (1)
 - What genetic disorder would result in humans if this abnormality occurred in chromosome pair no. 21? (1)
 - Give one symptom of the genetic abnormality mentioned in question 2.2.4 (b). (1)
- (8)

- 2.3 The graph shows information about the movement of chromatids in a cell that has just started Metaphase II.



- 2.3.1 Name one difference between Metaphase I and Metaphase II. (2)
- 2.3.2 What is the duration of Metaphase II in this cell? (1)
- 2.3.3 Use line X to calculate the duration of Anaphase II in this cell. (2)
- (5)
[25]

QUESTION 3

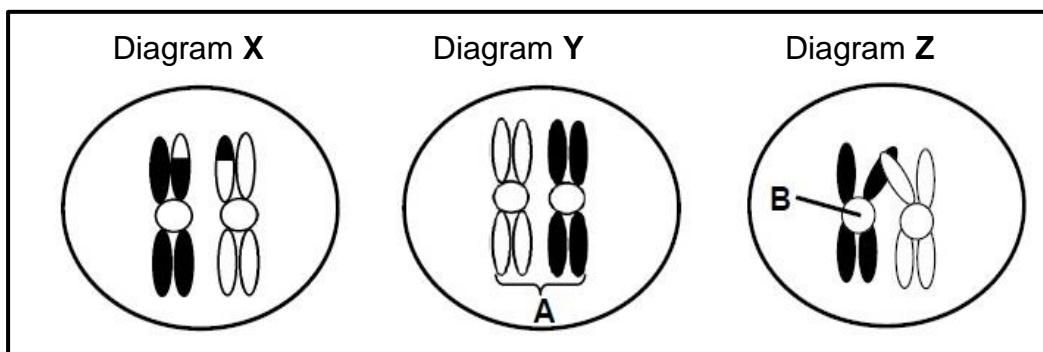
- 3.1 Describe the behaviour of the chromosomes during the process of meiosis I by referring to the following phases:
- 3.1.1 Prophase I (6)
- 3.1.2 Metaphase I (3)
- 3.1.3 Anaphase I (2)

3.1.4 Telophase I

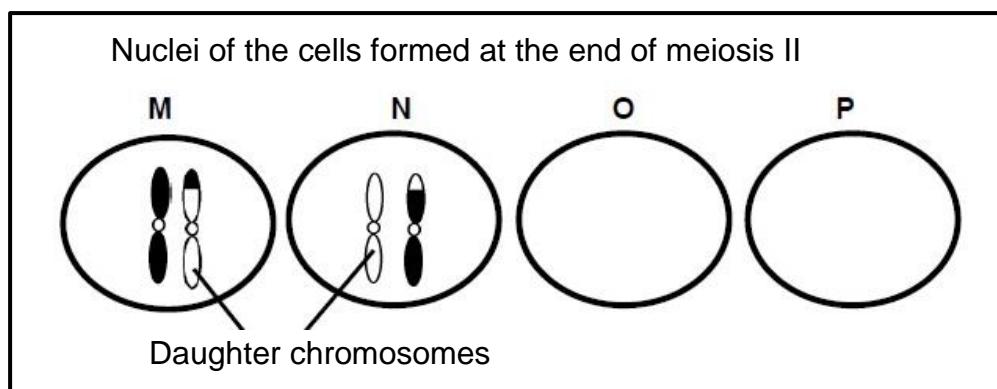
(3)

(14)

- 3.2 The diagram below shows chromosome pair 21 in the nucleus of a cell of the ovary of a woman. The chromosomes are involved in a process that takes place in a phase of meiosis.

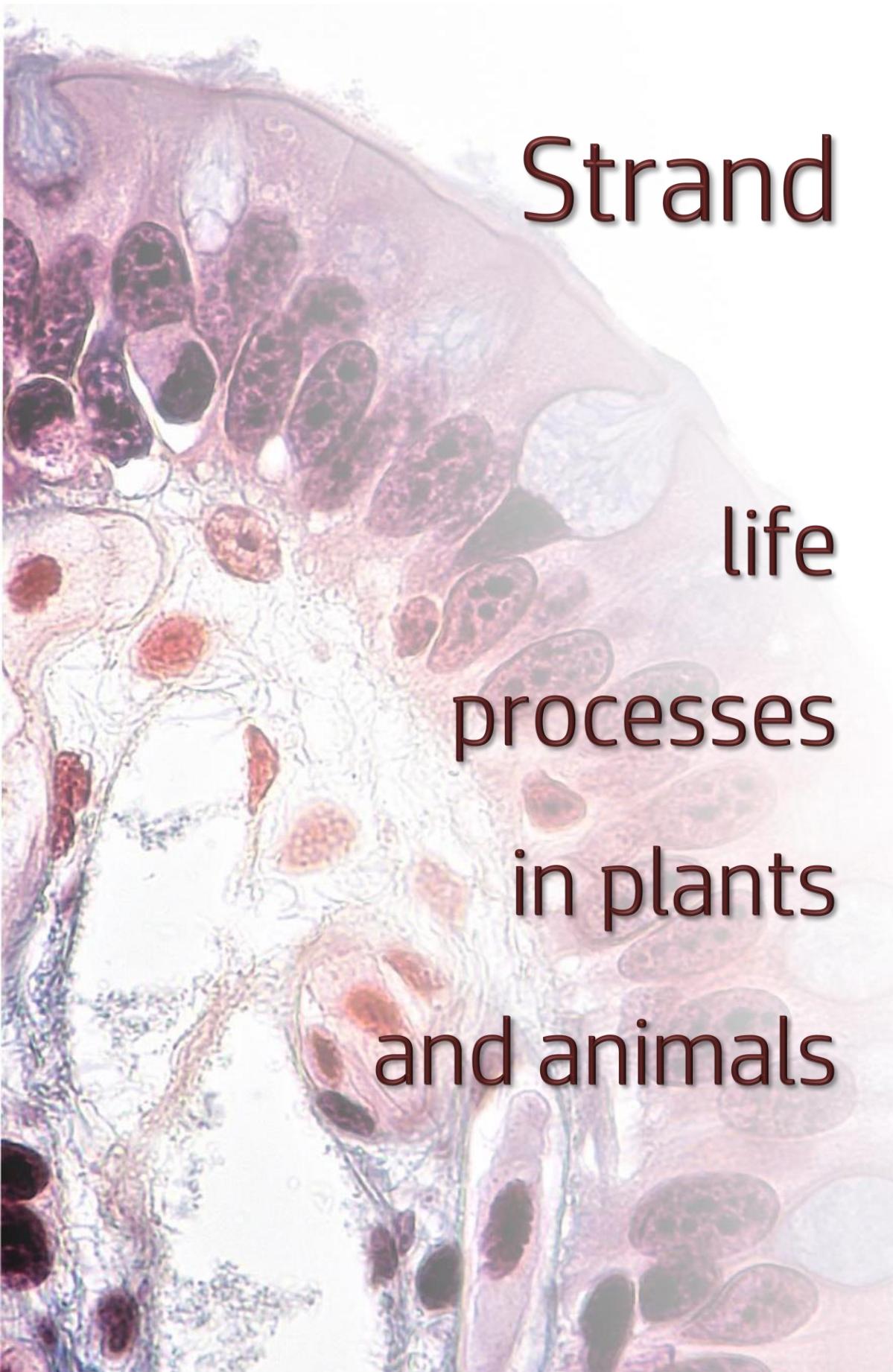


- 3.2.1 Give labels for **A** and **B**. (2)
- 3.2.2 Rearrange the letters **X**, **Y** and **Z** to show the correct sequence in which the events take place in this phase. (1)
- 3.2.3 Explain why chromosomes in Diagram X and Diagram Y are different in appearance. (3)
- 3.2.4 The diagram below shows the nuclei of the four cells that resulted from meiosis involving chromosomes in Diagram X above.



- a) Explain why nuclei **O** and **P** do NOT have chromosomes. (2)
- b) Name and explain the disorder that will result if Diagram **M** represents an egg cell that fuses with a normal sperm cell. (3)
- (11)

[25]**Section B: [50]****Total Marks: [100]**



Strand
life
processes
in plants
and animals

3: Reproductive strategies in vertebrates

Introduction

External and internal fertilisation

External fertilisation

Internal fertilisation

Comparison of external
vs internal fertilisation

Ovipary, ovovipary and vivipary

Comparison of ovipary,
ovovipary and vivipary

Activity 1:
Fertilisation

The amniotic egg

The amniotic egg consists of

Activity 2: Amniotic
egg

Precocial and Altricial
development

Major differences between
precocial and altricial
development

Parental care

Activity 3:
Development and
care

CHAPTER 3: REPRODUCTIVE STRATEGIES IN VERTEBRATES

Introduction

Reproduction ensures the continued existence of a species. Different species display different reproductive strategies to make sure that their offspring survive.

Reproductive strategies differ in:

- the number of eggs produced by the female
- the site of fertilisation, inside or outside the body of the female
- the place of development of the embryo and its nourishment
- how quickly the young can fend for themselves
- the type of parental care given to offspring.

Key terminology

reproductive strategy	structural, functional and behavioural adaptations that improve the chances of fertilisation and the survival of offspring
external fertilisation	fertilisation that takes place outside the female's body, usually in water
internal fertilisation	fertilisation that occurs inside the female's body where the male has deposited its sperm
ovipary	eggs are laid; the embryo develops outside the mother's body
ovoviviparity	young develop from eggs fertilised internally and retained within the mother's body after fertilisation until they hatch
viviparity	the young develop inside the uterus of mother after eggs are fertilised internally; young are nourished through the placenta
amniotic egg	the embryo inside the egg is protected by a hard shell; the egg consists of many extra-embryonic membranes that serve different functions
extra-embryonic membranes	membranes that surround the developing embryo inside the amniotic egg or uterus.
amnion	produces amniotic fluid which cushions embryo and protects it from mechanical injury, temperature changes, dehydration
allantois	collects the embryo's nitrogenous waste and assists in the exchange of gases
chorion	allows for gaseous exchange in the amniotic egg and forms the placenta in mammals

yolk sac	contains the food reserves for the developing embryo
precocial development	when hatchlings are well developed as they hatch, able to move and feed themselves, with eyes open – limited parental care
altricial development	when hatchlings are underdeveloped as they hatch, unable to move or feed or fend for themselves – young require more parental care
parental care	includes the building of nests, protection, teaching of young and feeding – the care, or lack thereof, directly influences the survival of the young

External and internal fertilisation

- Fertilisation occurs when a sperm cell and egg cell fuse to form a zygote.
- Fertilisation can either occur outside or inside the female's body and varies in its water dependency.

External fertilisation

External fertilisation takes place outside the female's body. Water is required for fertilisation (Figure 1A and 1B).



Figure 1A: Salmon breeding (spawning) in a lake



Figure 1B: Fertilised salmon eggs

Internal fertilisation

Internal fertilisation takes place inside the female's body. No water is required. See Figures 2 and 3.



Figure 2: Internal fertilisation in mammals via a penis



Figure 3: Internal fertilisation in birds

Comparison of external and internal fertilisation

Table 1 compares external with internal fertilisation.

Table 1: Comparison – external vs. internal fertilisation

External fertilisation	Internal fertilisation
• Requires water for fertilisation	• No water required for fertilisation
• Gametes (sperm and egg cells) are released into water	• Sperm cells are released into the female's body
• Many gametes released	• Fewer gametes released
• High mortality rates among young due to lack of protection. Eggs can easily desiccate or be predated on	• Lower mortality rates among young – protection provided by the mother's body or a hardened calcareous / leathery shell.
• e.g. fish and amphibia	• e.g. reptiles; birds and mammals

Ovipary, ovoviparity and viviparity

Ovipary, ovoviparity and viviparity are reproductive strategies that differ in respect to:

- where the zygote is formed
- where development occurs
- how the embryo receives its nourishment
- the type of egg or its presence or absence

Comparison of ovipary, oovoviparity and viviparity

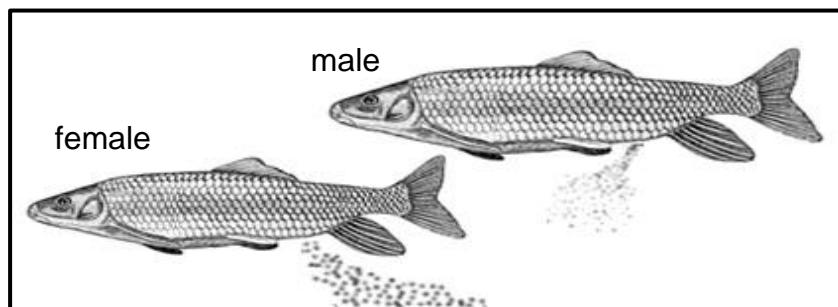
Table 2 compares the three reproductive strategies, namely, ovipary, oovoviparity and viviparity.

Table 2: Three reproductive strategies

	Ovipary	Oovoviparity	Vivipary
fertilisation	external or internal	internal	internal
development of embryo	external to the body of the female	inside the body of the female	inside the female's body
nutrition	Yolk is the only form of nutrition for the developing embryo and is usually present in small quantities	Yolk present in the egg. Young are independent of the mother's body	Young receive nutrition from the mother's body through the placenta
type of egg	jelly-like or calcareous	calcareous or leathery	None

Activity 1: Fertilisation

The diagram below shows a certain species of fish mating.



- Identify the type of fertilisation displayed by the fish species. (1)
- State two visible ways in which the chances of fertilisation in these fish are increased. (2)
- Name the reproductive strategy used by these fish that involves the production of eggs. (1)
- Give two reasons why there is no need for the eggs of these fish to be covered by a hard or leathery shell. (2)
- Explain the challenge that external fertilisation poses and how organisms with external fertilisation overcome this challenge. (4) (10)

The amniotic egg

The amniotic egg (Figure 4) is a major development in the evolution of animal life on land – from being water dependent for sexual reproduction, to being able to **reproduce without the availability of water**.

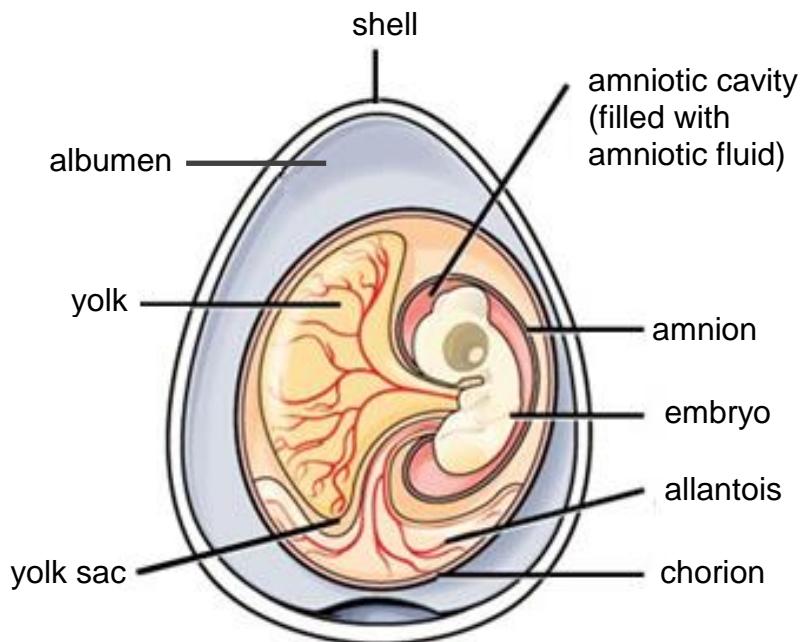


Figure 4: The amniotic egg

The amniotic egg consists of ...

- The **developing embryo**
- Three extra embryonic membranes:
 - the **amnion** – produces amniotic fluid which cushions the embryo and protects it against mechanical injury, temperature changes and dehydration
 - the **allantois** – collects nitrogenous waste and assists in the exchange of gases
 - the **chorion** – allows for gaseous exchange in reptiles and birds, where a shell is present and in mammals, where no shell is present, it forms the placenta.

- The **yolk sac**

The yolk sac contains the food reserves for the developing embryo. If yolk is present in smaller quantities, the young are hatched sooner, are under-developed and usually require more parental care. If yolk is present in larger quantities, the incubation period is longer, and the young are usually well developed when they hatch.

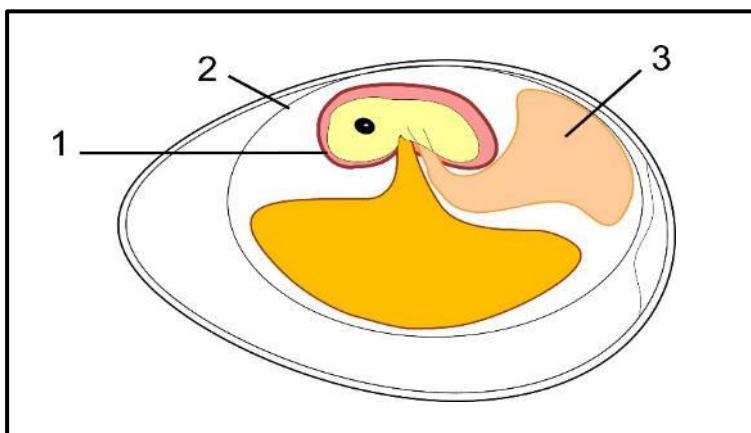
- A **hardened calcareous or leathery shell**

The shell helps to protect the developing embryo from mechanical injury and prevent desiccation, while still allowing gases to move through.

The amniotic egg: <https://youtu.be/Qq0kMEWzdHg>

Activity 2: Amniotic egg

Study the diagram and answer the questions that follow.



1. Identify the membrane numbered 1 and 2. (2)
2. Describe any two functions of the fluid found in part 1. (2)
3. Identify the organ that will replace the function of membrane 3 in the adult organism. (1)
4. Explain why the allantois and yolk sac are non-functional in a human foetus. (2)
5. Briefly explain how the amniotic egg allowed life to evolve onto land. (5)
(12)

Precocial and altricial development

Precocial and altricial development are terms used to describe how well-developed offspring are at birth.

Major differences between precocial and altricial development

(see Figures 5, 6, 7 and 8)

Table 3: Comparing precocial and altricial development

	Precocial development	Altricial development
Development of the body	well developed	under developed
Eyes after birth	open	closed
Presence of fur / feathers	have fur / feathers	usually naked
Parental care required	low degree of parental care required	high degree of parental care required
Mobility	young can move soon after birth	young have limited ability to move freely
Yolk amount in egg	greater quantity	lower quantity



Figure 5: Ducklings with down feathers and open eyes



Figure 6: Salmon fry with fins and open eyes



Figure 7: Nesting birds – note the eyes are closed and their bodies naked



Figure 8: 'Pinkies' – rat (mammal) babies – note no fur and eyes are closed

Parental care

In higher-order animals, parental care is a **behaviour that increases the survival of the young**. As a reproductive strategy, those animals which invest more energy pre-natally (before birth) usually display very little parental care once young have been born. In animals where less energy is invested pre-natally, more post-natal parental care is offered.

Parental care can be seen in the following examples:

- Building of nests and incubation of eggs
- Guarding from predators
- Teaching offspring

Examples of parental care: <https://youtu.be/7Ko07Md3XmU>

Activity 3: Development and care

Study the diagrams below showing different forms of development



A



B



C



D

1. Write down the letters of the organisms which show
 - a) altricial development (2)
 - b) precocial development (2)
2. Ovoviparous animals can display either precocial or altricial development. Explain how these development approaches differ in respect to
 - a) the degree of parental care offered (4)
 - b) how well young are developed at birth (2)
 - c) the amount of yolk present in eggs (2)
3. Tabulate three differences between precocial and altricial development. (7)
(19)

4: Human reproduction

Introduction	Uterine cycle
The male reproductive system	Hormonal control of the menstrual cycle
The female reproductive system	Activity 3: Hormones
Activity 1: Reproductive systems	Activity 4: Menstrual cycle
Puberty	Fertilisation and development of zygote to blastocyst
Gametogenesis	Implantation of the blastocyst and gestation
Spermatogenesis	Activity 5: Fertilisation
Oogenesis	End of topic exercises
Activity 2: Gametogenesis	
The menstrual cycle	
Ovarian Cycle	

CHAPTER 4: HUMAN REPRODUCTION

Introduction

In this chapter we will be studying human reproduction. All organisms must reproduce to ensure the survival of the species. We will look at the structure of the male and female reproductive systems, puberty, how gametes are produced, and hormonal changes that occur in the menstrual cycle. We will also briefly study fertilisation and the development of the zygote, including implantation and gestation.

The understanding of this topic necessitates understanding the role of meiosis, mitosis and fertilisation in the human life cycle as illustrated in Figure 1 below.

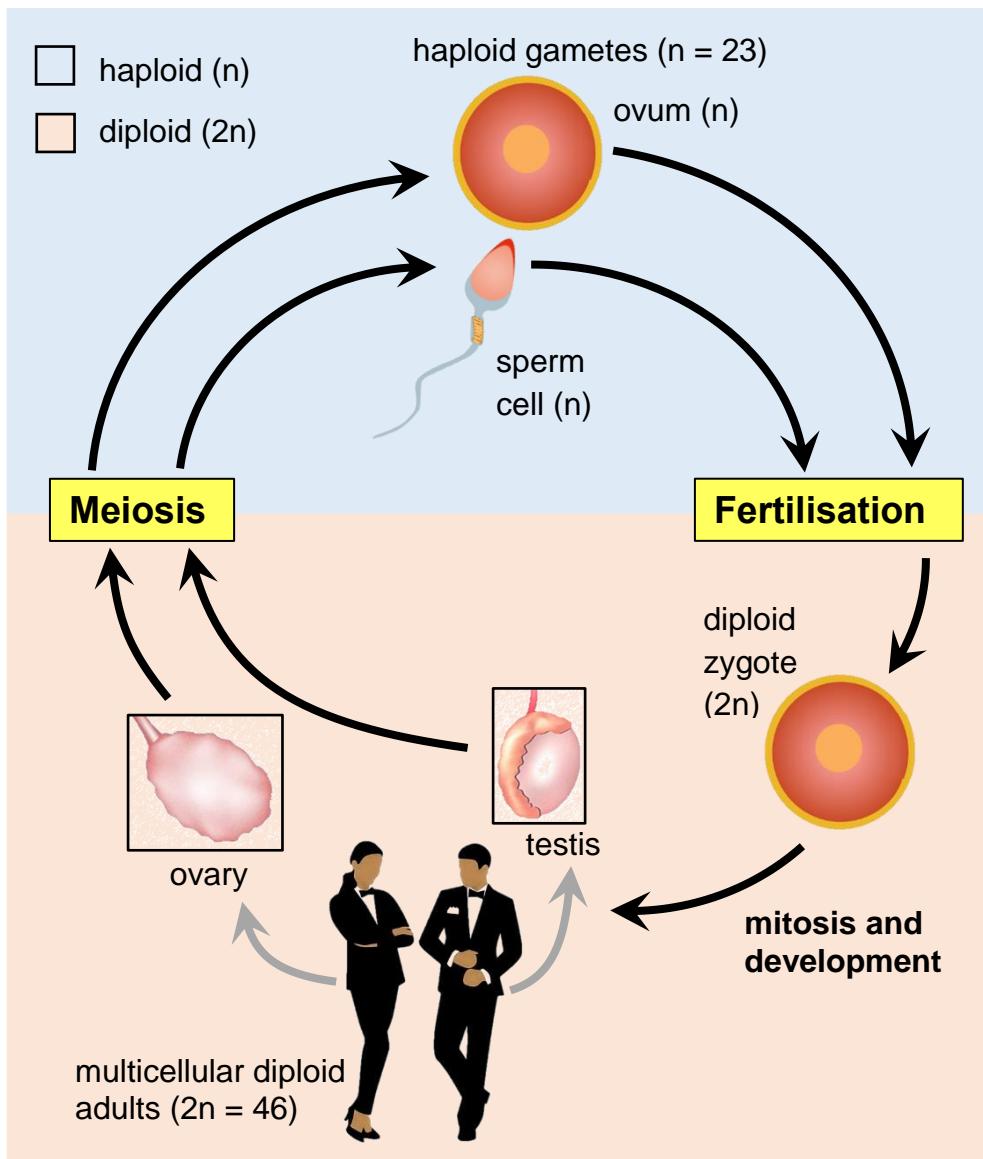


Figure 1: Human life cycle

All the body cells (somatic cells) of a human being are **diploid (2n)** i.e. have two sets of each chromosome. For humans to grow or to repair damaged tissues, the somatic cells divide by **mitosis**. The new cells produced by mitosis are identical to the original cells which divided.

Sexual reproduction requires two parents. Both the male and the female produce **gametes** (egg and sperm) by a reduction division referred to as **meiosis**. Meiosis ensures that the gametes are **haploid** i.e. they have only one set of chromosomes. When two gametes fuse (a sperm and an egg) as a result of **fertilization**, a diploid **zygote** is formed. The zygote then divides by mitosis to form a human.

Key terminology

gamete	an egg or sperm cell with half the number of chromosomes
gametogenesis	the process in which gametes are produced in the testes and ovaries through meiosis
oogenesis	the process that occurs when egg cells are made in the ovary through meiosis
spermatogenesis	the process that takes place when sperm cells are made in the testes through meiosis
germinal epithelium	cuboidal epithelium found on the surface of the testes and ovaries which gives rise to the cells which mature to form sperm cells and egg cells respectively

The male reproductive system

As shown in Figure 2 below, the male reproductive system consists of ...

- the main male sex organ – a pair of testes in the scrotum
- various ducts and tubules – seminiferous tubules, epididymis, vas deferens and the urethra
- accessory glands – prostate gland, Cowper's gland and seminal vesicles
- the external genitalia – penis

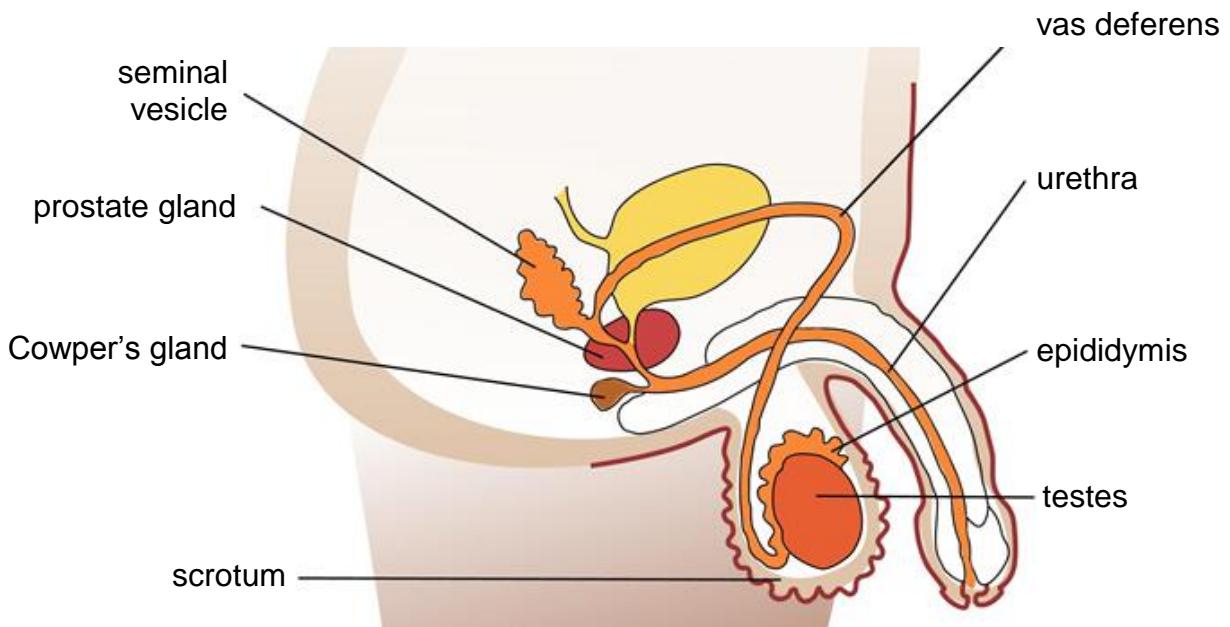


Figure 2: The male reproductive system

The functioning of each part of the reproductive system is explained in Table 1.

Table 1: Structure and function of the male reproductive system

Part	Structure	Function
testes	oval shaped glands, suspended in the scrotum	produce sperm cells and the hormone testosterone
scrotum	skin sac that holds the testes	protects the testes and holds the testes “outside” the body, at 2°C lower than body temp.
epididymis	coiled tubule on the outside of the testes but still in the scrotum	temporarily stores spermatids until they mature into sperm cells
vas deferens	muscular tube passing from the epididymis to the urethra	transports sperm from the epididymis to the urethra
urethra	tube which runs through the penis	transports urine and semen out of the body
prostate gland	gland found below the bladder, at the point where the urethra begins; the largest accessory gland	produces a nutrient-rich fluid that provides energy for the sperm cells
Cowper's glands	small pair of glands found below the prostate gland.	produces mucus that helps with the movement of sperm cells
seminal vesicles	medium sized pair of glands attached to the end of the vas deferens	produces alkaline fluid to neutralise vaginal acids which would kill sperm

The testes contain **seminiferous tubules** (see Figure 3).

- The tubules are lined by germinal epithelium cells which produce sperm cells.
- Some of the cells develop into **Sertoli cells** which provide nutrients for the spermatids to become mature sperm cells. The seminiferous tubules are surrounded by connective tissue that contain the **Cells of Leydig** which produce testosterone.

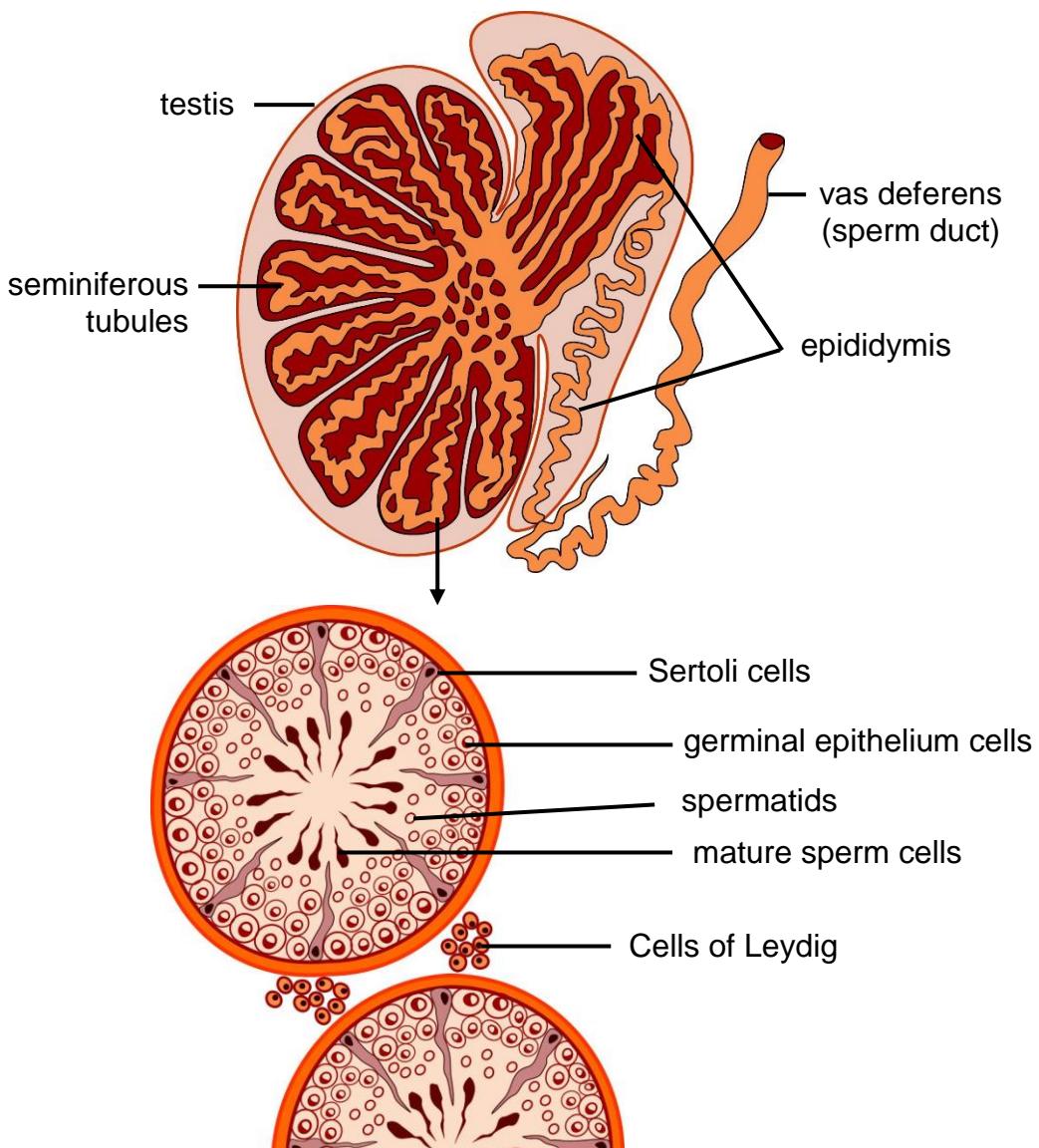


Figure 3: Cross-section through the seminiferous tubules of the testes

Testosterone has the following functions:

- development of the male secondary sexual characteristics
- stimulating the maturation of sperm cells

It is vitally important that the testes are suspended on the outside of the body, as this allows for temperature regulation to occur. The optimum temperature for sperm production is 2 to 3°C lower than normal body temperature.

If the temperature in the scrotum is high, it interferes with the quality of the sperm resulting in male infertility.

By having the testes suspended in the scrotum, the temperature of the testes can be adjusted by moving the testes closer to the body in cold conditions or further away from the body during warm conditions.

The male reproductive system: <https://www.youtube.com/watch?v=k60M1h-DKvY>

The female reproductive system

The female reproductive structure consists of:

- the main female sex organ – the ovaries
- the ducts – fallopian tubes
- the accessory organs – the uterus and the vagina
- the external genitalia – the vulva

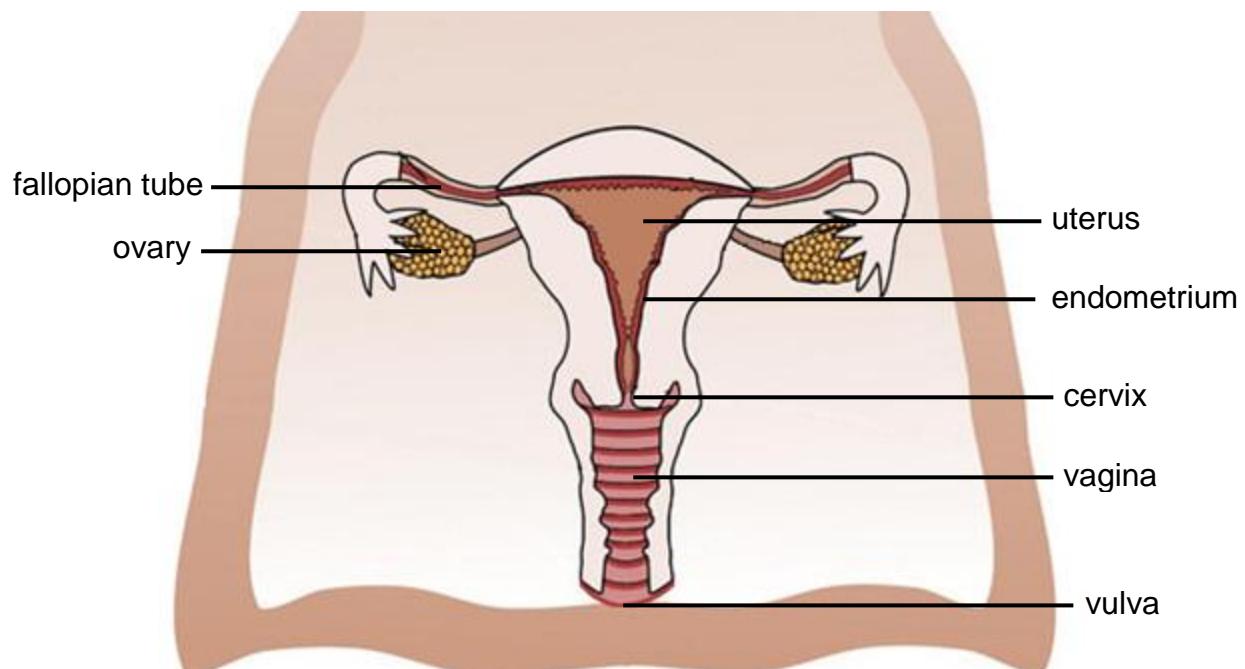


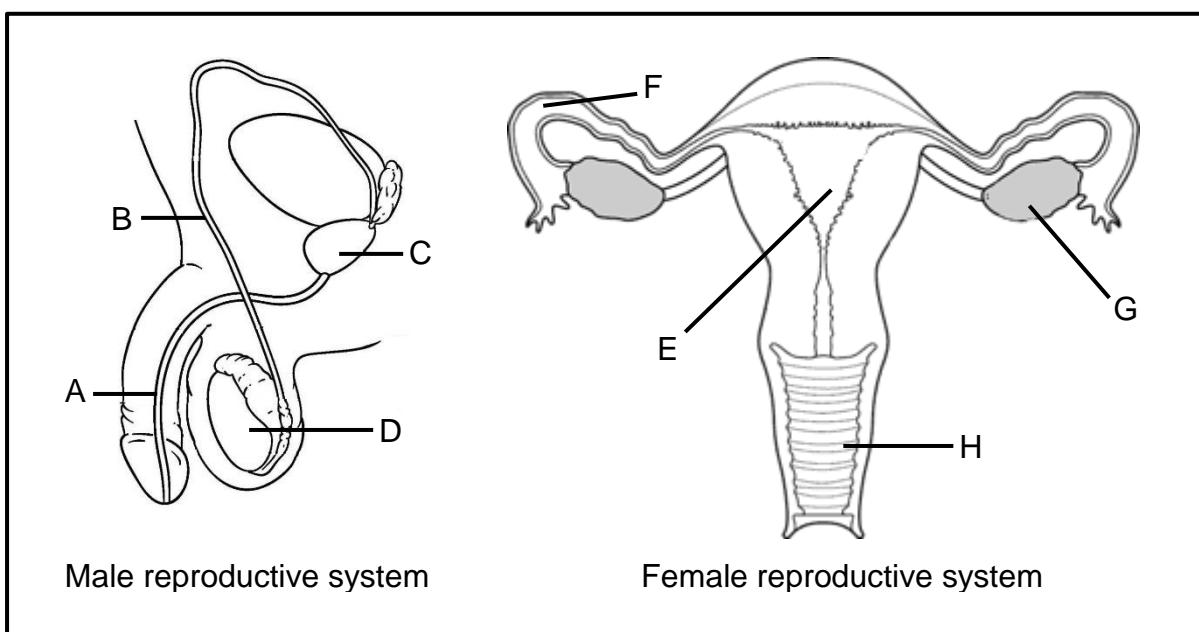
Figure 4: The female reproductive system

Table 2: Structure and function of the female reproductive system

Part	Structure	Function
ovaries	found as a pair, one on either side of the uterus, and surrounded by germinal epithelium	produce egg cells, secrete the hormones progesterone and oestrogen
fallopian tubes	connect the ovaries to the uterus – are lined with ciliated columnar epithelium which helps the movement of the egg cells	transports egg cells from the ovary to the uterus; the site of fertilisation
uterus	hollow, pear-shaped organ	houses and protects the embryo and foetus during pregnancy
endometrium	inner lining of the uterus	site of implantation and where the placenta forms
cervix	lower, narrow opening of the uterus	stretches and opens to allow the baby through during childbirth
vagina	muscular tube which runs from the cervix to the exterior	receives the penis and semen during sexual intercourse; the birth canal; passage for menstrual blood
vulva	opening to the vagina; covered by two vaginal covers called the labia	protects the entrance to the vagina

Activity 1: Reproductive systems

Study the diagrams below showing the male and female reproductive systems.



- Identify the parts labelled A – H. (8)
 - State one function of each of the following:
 - The fluid produced by C. (1)
 - Part E (1)
 - Provide two functions of part H. (2)
 - Explain why it is necessary for part D to be ‘outside’ the human body in males. (2)
- (14)

Puberty

Puberty is the period during which males and females reach sexual maturity. Puberty usually begins between the ages of 11 to 15, though it may occur much earlier or later depending on the individual. During puberty the sex hormones are produced which stimulate gametogenesis and sexual maturity. At the same time secondary sexual characteristics develop. Table 3 below lists the various changes in the development of males and females that take place during puberty.

Table 3: The development of males and females during puberty

Males	Females
male sex hormone testosterone is produced	female hormones oestrogen and progesterone are produced
growth of hair around the scrotum (pubic hair)	growth of hair around the vulva (pubic hair)
growth of hair in the armpits	growth of hair in the armpits
growth of hair on the face	
larynx enlarges / voice becomes deeper	
muscles enlarge and the shoulders become wider	the hips become wider and fat is deposited below the skin
penis and the testes enlarge	development of breasts

Gametogenesis

Gametogenesis is the term used to describe the process by which gametes are produced from the germinal epithelium in the sex organs.

It includes spermatogenesis and oogenesis.

Spermatogenesis

Spermatogenesis is the production of male gametes (sperm cells) in the testes of the male. It occurs in the germinal epithelium of the seminiferous tubules in the testes. This process happens under the influence of testosterone. During puberty the germinal epithelium contains a diploid number of chromosomes (46). These cells go through the process of meiosis forming haploid sperm cells with 23 chromosomes. The gametes may have (22 + X) or (22 + Y) chromosomes.

Spermatogenesis takes place as follows:

- Under the influence of testosterone, the diploid germinal epithelial cells ($2n$) lining the seminiferous tubules go through meiosis
- Each cell that goes through meiosis produces 4 haploid spermatids (n)
- Each spermatid matures to form a haploid sperm cell (see Figure 3 above)

Figure 5 below shows the structure of a human sperm cell. Each sperm cell is made up of a head, middle portion (neck) and a long tail.

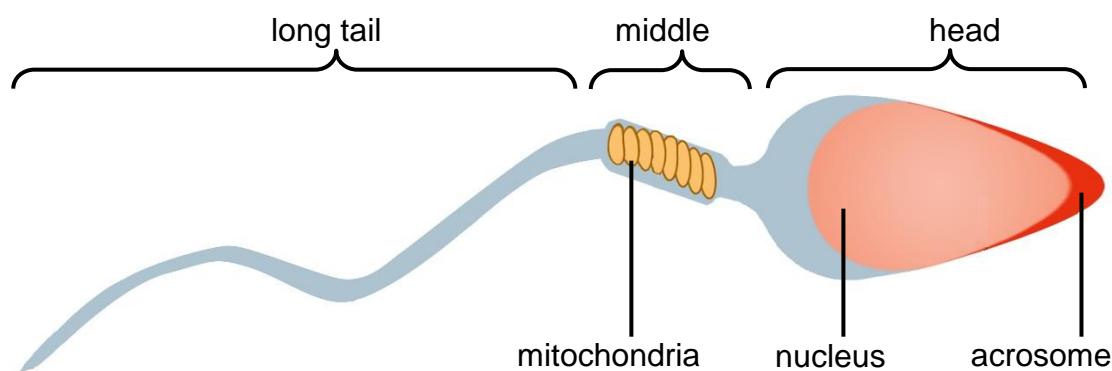


Figure 5: The structure of a sperm cell

- The head is mostly made up of the nucleus which contains 22 autosomes and one sex chromosome (X or Y).
- The acrosome (also in the head) contains enzymes that dissolve the outer layer of the egg allowing fertilisation to occur.
- The middle portion contains mitochondria which provide energy for the movement of the sperm cell.
- The long tail allows the sperm cell to propel itself forward (to swim) through fluid.

Oogenesis

Oogenesis is the production of female gametes (ova / egg cells) in the ovaries of a female. It occurs when the diploid germinal epithelium of the ovaries starts to produce follicles by mitosis.

Oogenesis takes place as follows:

- The diploid germinal epithelium cells ($2n$) of the ovaries go through the process of mitosis to form many follicles
- Every 28 days, the follicle stimulating hormone (FSH) stimulates one follicle. Only one cell inside of that follicle enlarges and goes through the process of meiosis
- Out of the 4 (four) haploid cells produced through meiosis, only one cell will survive to form a mature ovum
- The other three cells from meiosis will degenerate

Figure 6 shows the structure of a human egg (ovum). Each ovum is made up of follicle cells, a layer of jelly, cytoplasm and a haploid nucleus.

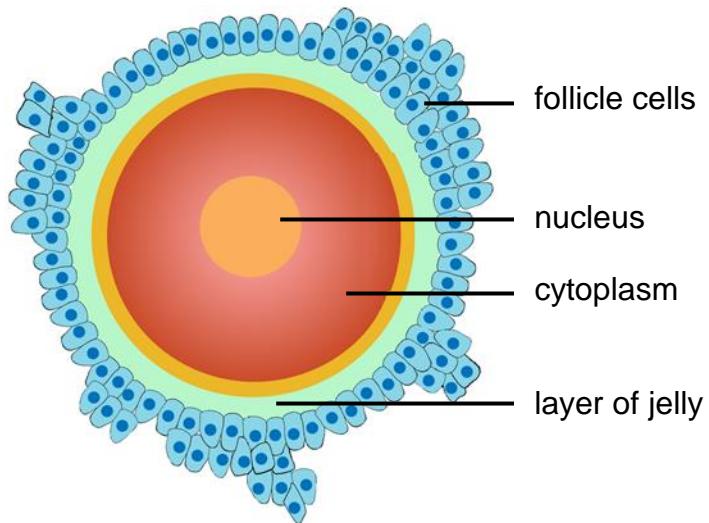


Figure 6: The structure of an ovum

- The nucleus contains 22 autosomes and one sex chromosome (X)
- The cytoplasm nourishes the egg
- The jelly layer provides protection for the early developmental stages of the fertilised egg

The structure of sperm and egg cells:

<https://www.youtube.com/watch?v=CuxaXghfyE&list=PLW0gavSzhMIQYSpKryVcEr3ERup5SxHI0&index=43>

Activity 2: Gametogenesis

1. Name the organ where meiosis takes place in the male and female reproductive systems respectively. (2)
2. Define gametogenesis. (1)
3. Name the type of gametogenesis that takes place in the male and female reproductive systems respectively. (2)
4. Draw a fully labelled diagram of an ovum. (5)
5. Discuss the functions of the four main parts of a sperm cell. (8)
(18)

The menstrual cycle

Key terminology

Graafian follicle	mature follicle inside the ovary filled with fluid in which the ovum grows
ovulation	the release of an ovum from the Graafian follicle of the ovaries
endometrium	the inner lining of the uterus wall
menstruation	the monthly loss of blood and tissue as a result of changes that occur in the lining of the uterus
menopause	stage in the life of a woman when she stops ovulating and menstruating; usually occurs between the ages of 45 and 55
fertilisation	the fusion of the haploid sperm cell nucleus and the haploid egg cell nucleus to form a diploid nucleus of the zygote
implantation	the attachment of the embryo to the endometrium lining the uterus

The menstrual cycle refers to changes that occur in the ovaries and uterus of a female over a period of 28 days. This cycle begins at puberty and ends at menopause.

The menstrual cycle is made up of two separate cycles that happen at the same time:

1. Ovarian cycle
2. Uterine cycle

1. Ovarian cycle

The ovarian cycle refers to the development and release of an ovum (or egg cell). This takes place inside the ovary. The ovarian cycle begins when FSH (Follicle Stimulating Hormone) is secreted by the pituitary gland. FSH is transported to the ovary by the blood. The following diagram (Figure 7) illustrates the ovarian cycle.

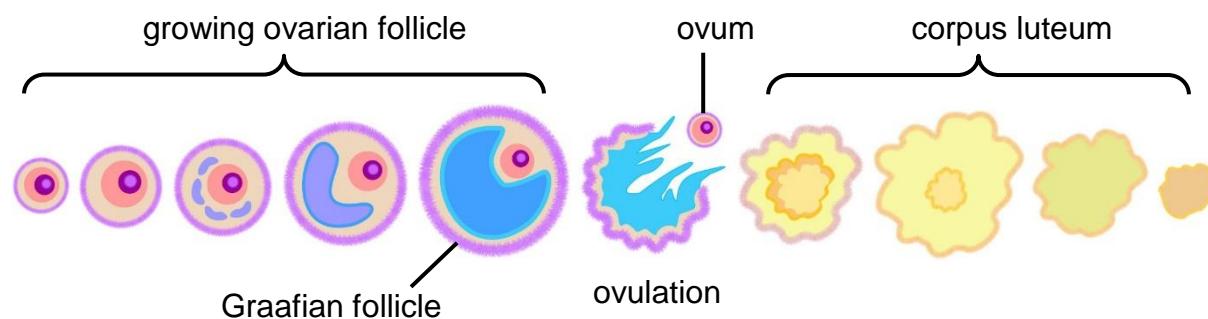


Figure 7: The ovarian cycle

1. FSH stimulates a primary follicle to become a **Graafian follicle** which contains a mature ovum (or egg cell).
2. As the Graafian follicle develops, it produces the hormone **oestrogen**, increasing the oestrogen levels in the blood.
3. Around Day 14, the Graafian follicle ruptures and releases an ovum in a process called **ovulation**. Ovulation is stimulated by the Luteinising Hormone (LH) which is released by the pituitary gland.
4. LH causes the ruptured Graafian follicle to change into a structure called the **corpus luteum**. The corpus luteum secretes the hormone **progesterone** increasing the levels of progesterone in the blood.

- If fertilisation does not take place, the corpus luteum shrinks and stops producing progesterone. The ovum passes down the fallopian tube, enters the uterus and leaves the body through menstruation.

NOTE:

- If the ovum is fertilised, the corpus luteum remains active and continues secreting progesterone
- Oestrogen and progesterone produced by the ovaries during the ovarian cycle influence the uterine cycle

2. Uterine cycle

The Uterine cycle shows the changes that occur in the uterus wall as it gradually thickens and becomes more vascular (richly supplied with blood vessels) over a period of 28 days.

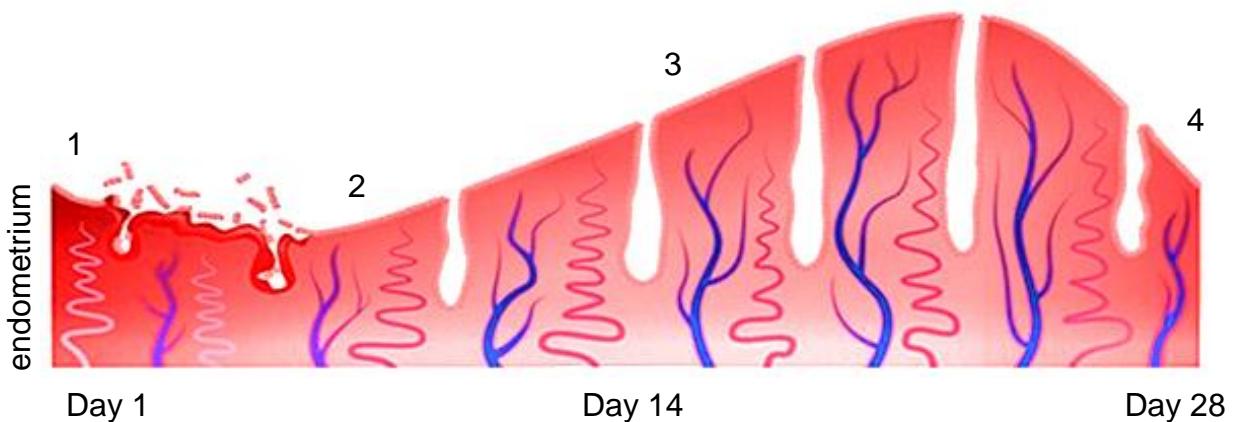


Figure 8: Illustrating the changes to the endometrium

- The endometrium breaks down and is released (menstruation). This lasts for approximately 4 to 7 days.
- The endometrium is stimulated by oestrogen to become thicker and develop more blood vessels and glands.
- Progesterone stimulates the endometrium to become even thicker and develop more blood vessels and glands. This happens in preparation for possible implantation of the fertilised ovum.
- If fertilisation does not take place, the endometrium tears away resulting in menstruation.

Figure 9 and Table 4 below summarise the changes that occur in the ovarian and uterine cycles over a period of 28 days (the menstrual cycle)

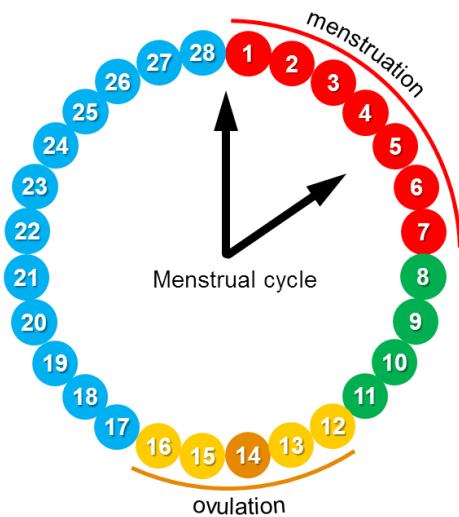


Figure 9: The menstrual cycle

Table 4: A summary of changes in the ovarian and uterine cycles

Days	Ovaries	Uterus
1 – 7	new follicles develop and secrete oestrogen	lining breaks down and is released (menstruation)
8 – 13	mature Graafian follicle develops and secretes oestrogen	oestrogen stimulates the endometrium to become thicker, more glandular and vascular
14	Graafian follicle bursts to release an ovum (ovulation)	
15 – 22	Graafian follicle becomes the corpus luteum which secretes progesterone	progesterone stimulates the endometrium to become even thicker, more glandular and more vascular to receive a fertilised ovum
23 – 28	no fertilisation: the corpus luteum shrinks and stops producing progesterone with fertilisation: corpus luteum remains active and continues producing progesterone no more follicles develop no menstruation takes place	

The menstrual cycle:

<https://www.youtube.com/watch?v=VI2wRbO8LZU&list=PLW0gavSzhMIQYSpKryVcEr3ERup5SxHI0&index=11>

Hormonal control of the menstrual cycle

The menstrual cycle is controlled by hormones. The ovarian cycle is influenced by follicle stimulating hormone and luteinizing hormone while the uterine cycle is influenced by oestrogen and progesterone. The levels of the hormones change during the different stages of the menstrual cycle and have an influence on each other.

The graph in Figure 10 below shows the changes in the levels of the individual hormones during the menstrual cycle.

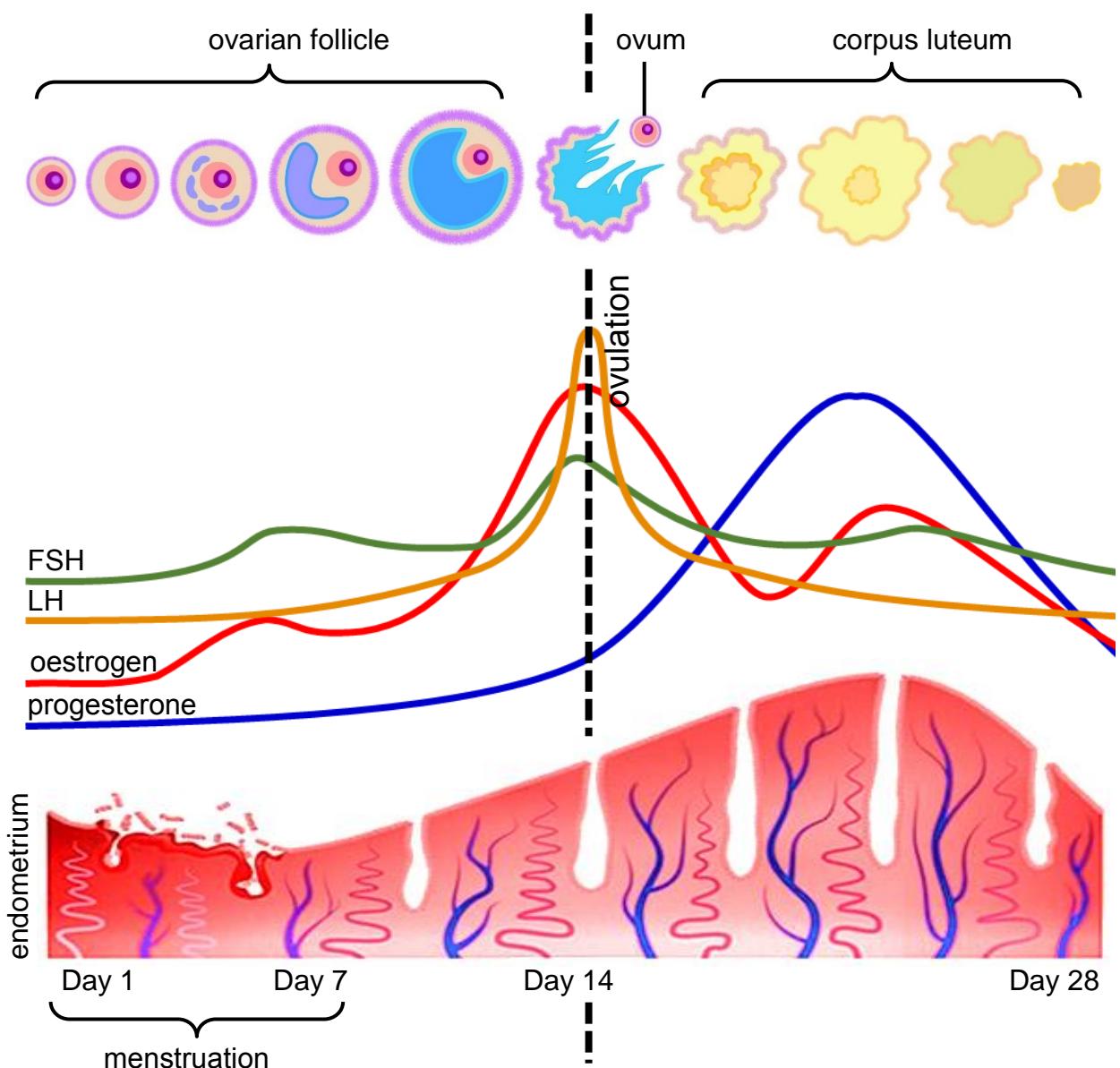


Figure 10: Hormonal control of the menstrual cycle

NOTE: This graph often appears in examinations. Make sure that you can interpret and understand what is happening to the levels of the different hormones and how they influence each other.

The hormonal control of the menstrual cycle takes place as follows:

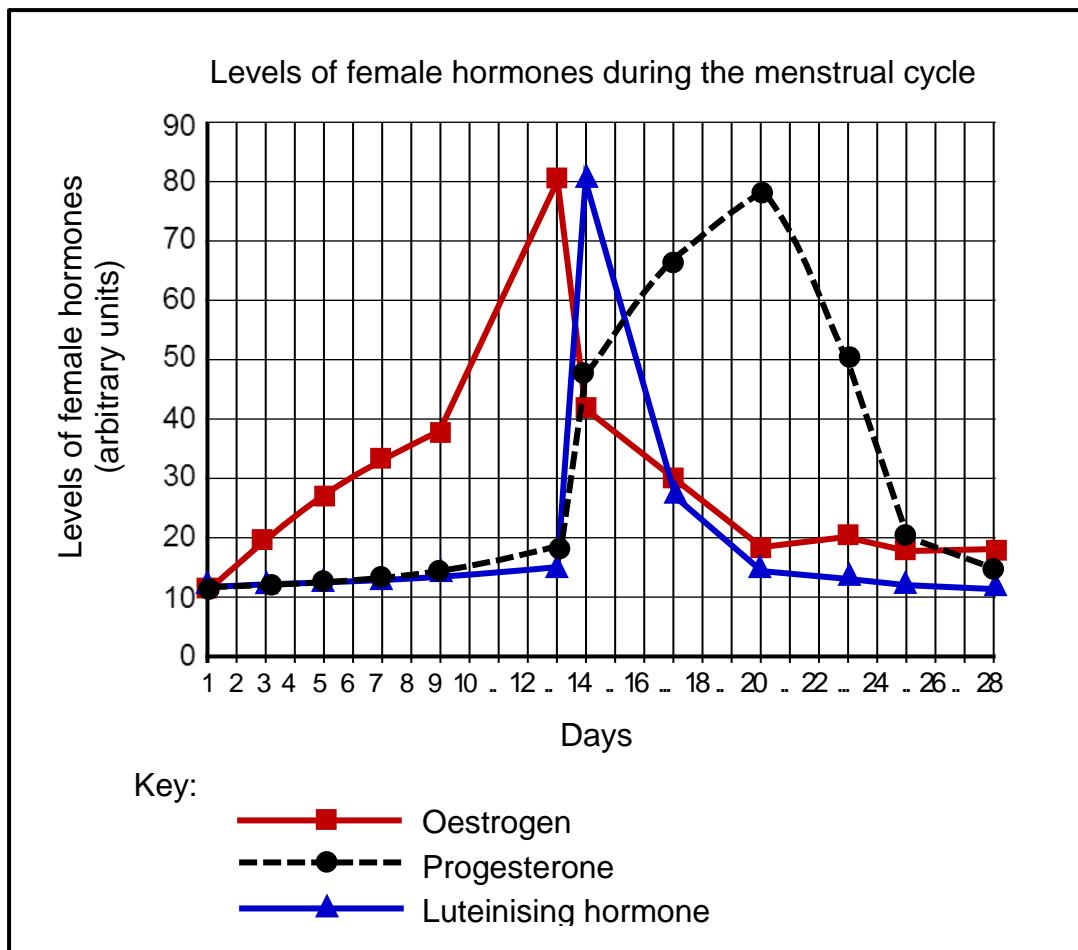
- Follicle stimulating hormone (FSH) released by the pituitary gland stimulates the development and maturation of a primary follicle in one of the ovaries
- As the follicle develops into a mature Graafian follicle it releases oestrogen
- The increasing oestrogen levels stimulate the pituitary gland to release luteinising hormone (LH)
- The increase in luteinising hormone (LH) causes ovulation to occur
- After ovulation occurs the Graafian follicle is changed into the corpus luteum which secretes progesterone
- The increased amount of progesterone prevents the release of follicle stimulating hormone and luteinising hormone (it inhibits them)
- As the corpus luteum breaks down the level of progesterone decreases, causing the endometrium to break down
- The endometrium and unfertilised ovum are released through the vagina as blood during menstruation
- Due to the decreased level of progesterone, the follicle stimulating hormone and the luteinising hormone are no longer inhibited. They are produced by the pituitary gland and the cycle begins again

Negative feedback mechanism between progesterone and FSH

A negative feedback system occurs in the menstrual cycle. A negative feedback mechanism is an interaction between two hormones, where an increase in one hormone stimulates an increase in the other hormone, which inhibits the first hormone, thus restoring balance. The negative feedback system can be seen in the hormonal control of the menstrual cycle where progesterone influences the secretion of follicle stimulating hormone. If the ovum is fertilised, the corpus luteum remains active and continues secreting progesterone. Increased levels of progesterone in the blood inhibit the secretion of the follicle stimulating hormone. As a result, no further development of the follicle occurs. Ovulation does not take place.

Activity 3: Hormones

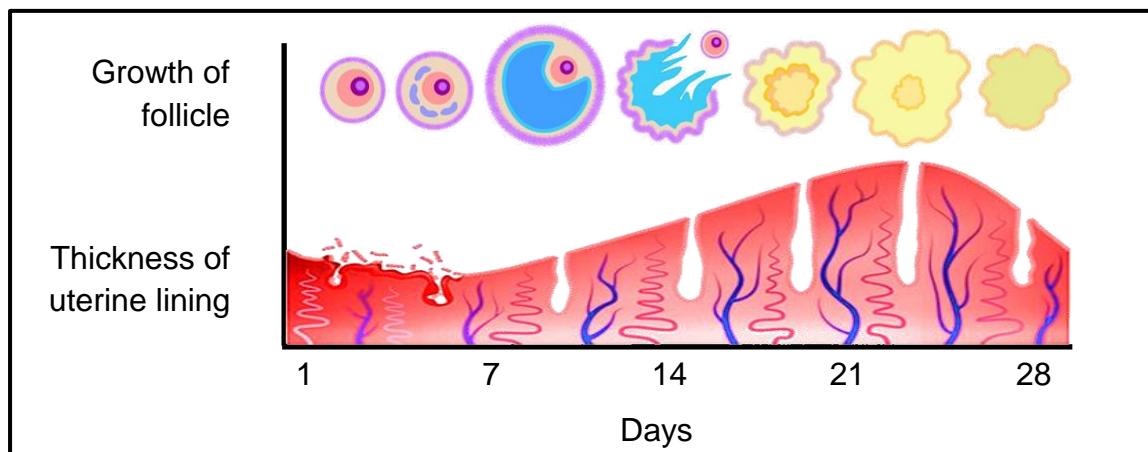
Study the graphs below, then answer the questions that follow.



- On which day did ovulation occur? (1)
- Give one reason for your answer to question 1 that can be seen on the graph. (1)
- Which structure in the ovary produces the following hormones?
 - Oestrogen (1)
 - Progesterone (1)
- Explain why there is a sharp increase in the production of ...
 - oestrogen from day 9 to 13. (2)
 - luteinising hormone from day 13 to 14. (2)
- What conclusion can be drawn if the level of progesterone ...
 - Remains high from day 20 to 28? (1)
 - Drops as shown in the graph above? (1)(10)

Activity 4: Menstrual cycle

The diagram shows some of the changes that may take place during the menstrual cycle.



1. The menstrual cycle is controlled by hormones. Name one hormone that will increase in level between days 2 and 10. (1)
 2. Give one observable reason for your answer to question 1. (2)
 3. Explain what evidence there is in the diagram to indicate that no fertilisation took place? (3)
- (6)

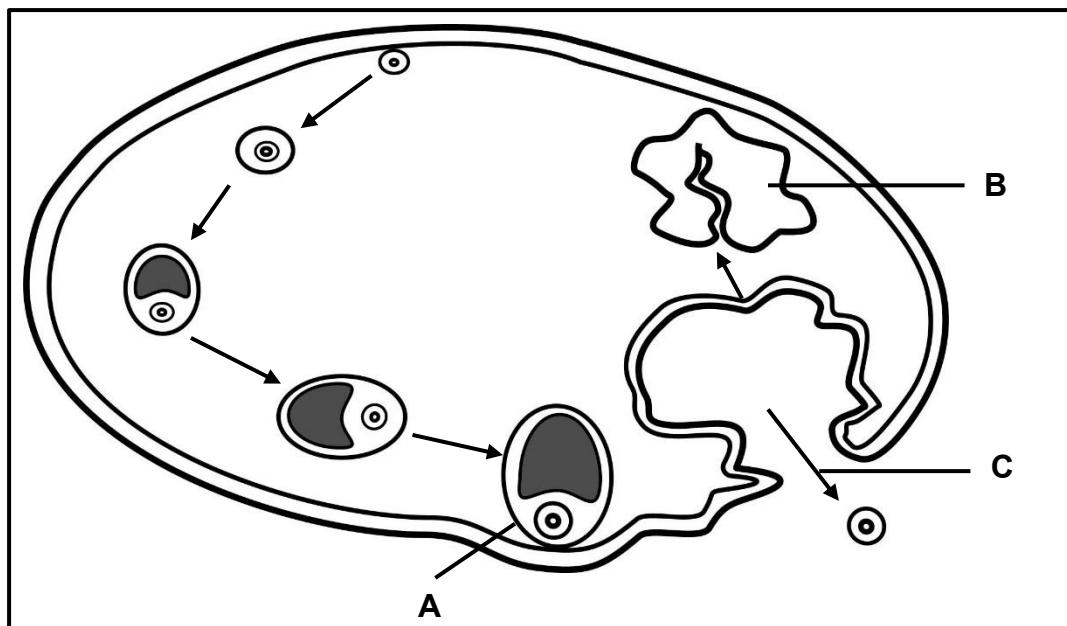
Fertilisation & development of zygote to blastocyst

During copulation (sexual intercourse) the penis is inserted into the vagina and sperm cells are released through ejaculation close to the cervix. The sperm cells swim through the cervix up into the uterus and through the fallopian tubes.

The haploid ovum released during ovulation enters the fallopian tubes. If an ovum (haploid) is present in the fallopian tubes, one sperm cell (haploid) may penetrate through the jelly layer and fertilise the ovum resulting in a diploid zygote. The nucleus of the ovum and the nucleus of the sperm cell fuse resulting in fertilisation.

The zygote divides by mitosis as it moves down the fallopian tube towards the uterus. Mitosis continues and a solid ball of cells known as the morula is formed. The morula develops into a hollow fluid-filled ball of cells called the blastocyst. Once the ovum is fertilised it takes approximately 5 days to form the blastocyst.

- 2.2. The diagram below represents the sequence of events that takes place during the ovarian cycle of a female.



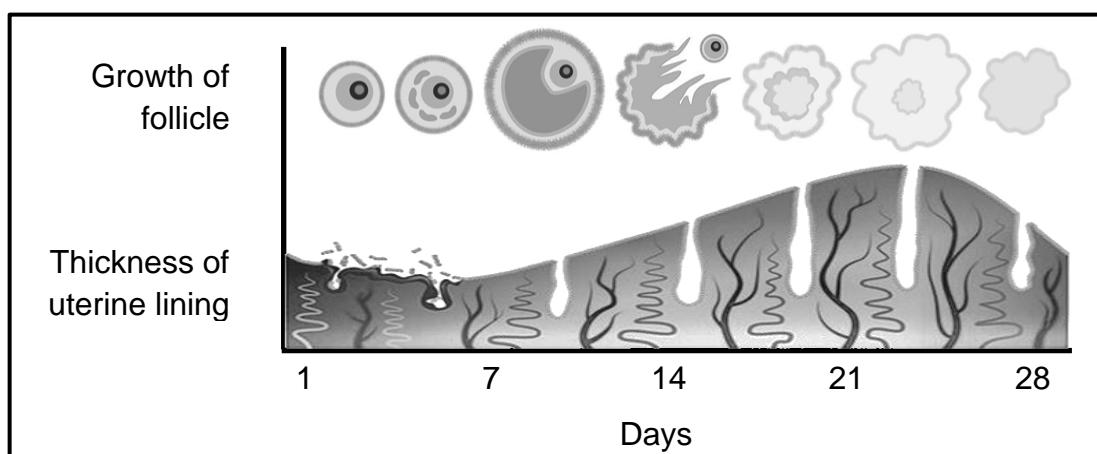
2.2.1 Give the name of the:

- a) Hormone that controls the development of structure A. (1)
- b) Process taking place at C. (1)

2.2.2 Structure B degenerates if fertilisation does not take place. Explain the implications of this for the ...

- a) ovarian cycle (2)
 - b) uterine cycle (2)
- (6)

2.3. The diagram shows some changes during the menstrual cycle.

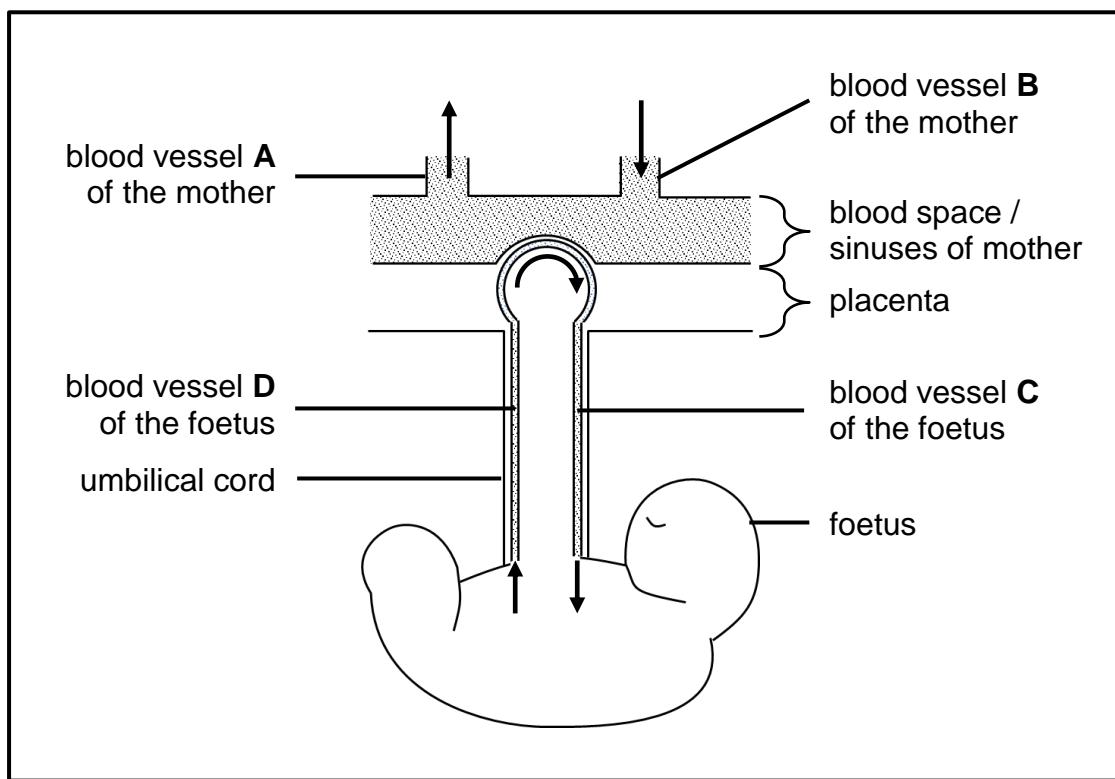


- 2.3.1 Describe the developmental changes in the fertilised ovum until implantation occurs in the uterus. (4)

- 2.3.2 Some females use an ovulation monitor so that they can be aware of the days when they are fertile. These monitors measure the level of hormones in the blood.
- Why would females want to know when they are fertile? (1)
 - Explain which hormone is likely to be monitored by the ovulation monitor. (3)
- (8)
[25]

Question 3

- 3.1 The diagram below represents the relationship between the blood system of the foetus and that of the mother. The arrows indicate the direction of blood flow in the blood vessels.



- Apart from playing a role in the diffusion of substances from the mother's blood to the foetus' blood, and vice versa, state two other functions of the placenta. (2)
- Blood vessel D is an artery. Tabulate two differences between the composition of the blood found in blood vessel C and blood found in blood vessel D. (5)
- Explain one consequence for the foetus if blood vessel D becomes blocked preventing blood flow. (2)

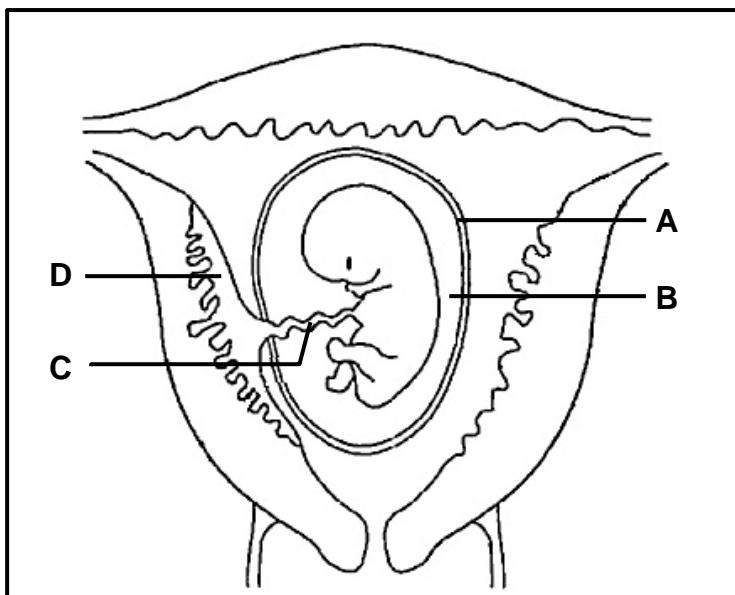
- 3.1.4 If the blood of the mother and the blood of the foetus come into contact with each other, it could lead to the death of the foetus. Describe why this would occur. (2)
(11)

- 3.2 Read the following extract and answer the questions that follow.

Several recent studies have suggested a gradual decline in sperm production in men. Endocrine disruptions as well as life style have been suggested as risk factors. One life style factor that may affect human fertility is driving a vehicle for a prolonged period. It is suggested that the driving position may increase the scrotal temperature.

- 3.2.1 State any one risk factor identified by the researchers. (1)
3.2.2 Explain why regular long-distance driving with no breaks could possibly lower the sperm count in healthy males. (3)
3.2.3 Suggest a consequence of lower sperm count in males. (2)
3.2.4 State any one daily life style trend or routine (other than the one mentioned in the extract) that should be avoided to maintain the optimum scrotal temperature. (1)
(7)

- 3.3 The diagram below represents a developing foetus in a human body.



- 3.3.1 Identify:
a) A (1)
b) C (1)
3.3.2 State two functions of the fluid B. (2)

- 3.3.3 Name one system in the baby's body that takes over the function of part **D** once the baby is born. (1)
- 3.3.4 Explain one negative impact on the foetal development if part **D** is reduced significantly. (2)
(7)
[25]

Section B: [50]

Total marks: [100]



Strand
diversity
change
and
continuity

5: Genetics and inheritance

Introduction	Multiple alleles e.g. blood grouping
Concepts of inheritance	Activity 5: Monohybrid crosses using blood types
Traits, genes and alleles	Dihybrid crosses
Activity 1: Traits, genes and alleles	Activity 6: Dihybrid crosses
Mendel as father of genetics	Genetic lineage (pedigrees)
Mendel's experiments	Activity 7: Pedigrees
Mendel's Laws of Inheritance	Mutations
Genetic diagrams	Gene mutations
Genetic crosses	Chromosomal Aberrations
Monohybrid crosses with complete dominance	Biotechnology
Activity 2: Monohybrid crosses with complete dominance	DNA profiling
Monohybrid crosses with incomplete dominance	Genetic engineering
Activity 3: Monohybrid crosses with incomplete dominance	Disadvantages of GMO's
Monohybrid crosses with co-dominance	Stem cell technology
Sex determination	Cloning
Sex-linked Inheritance	Advantages of cloning
Activity 4: Sex-linked diseases	Activity 8: Biotechnology
	Mitochondrial DNA and tracing genetic links
	Enrichment videos
	End of the topic exercises

CHAPTER 5: GENETICS AND INHERITANCE

Introduction

This section will build on what you have learnt about DNA, protein synthesis, meiosis and sexual reproduction and to help you understand how genetic characteristics e.g. physical structure, is passed down from generation to generation.

- Genetics is the study of heredity – how genetic characteristics are passed on from parents to child.
- Every individual inherits a set of genes found in chromosomes from a father and a mother which is unique to that individual but similar enough to identify the individual's species.

Key terminology

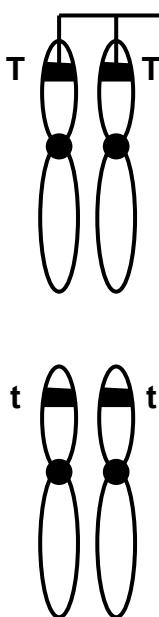
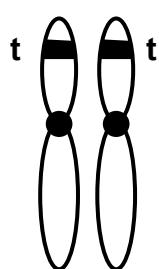
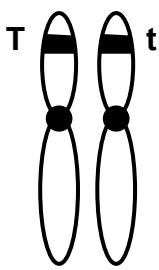
hereditary	passing of hereditary characteristics from parent to offspring
filial generation (F_1)	offspring of parent organisms
locus	the exact position (location) of a gene on a chromosome
genetic engineering	techniques used to change the genetic material of a cell or living organism – a form of biotechnology

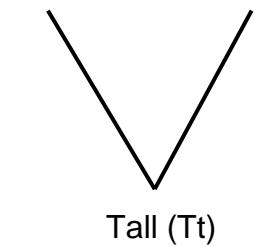
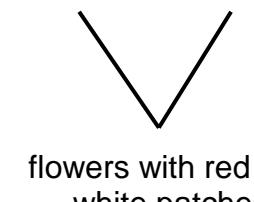
Concepts in inheritance

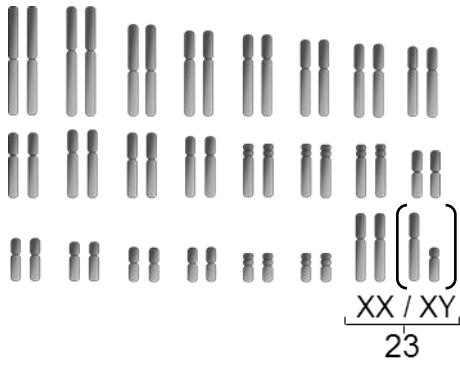
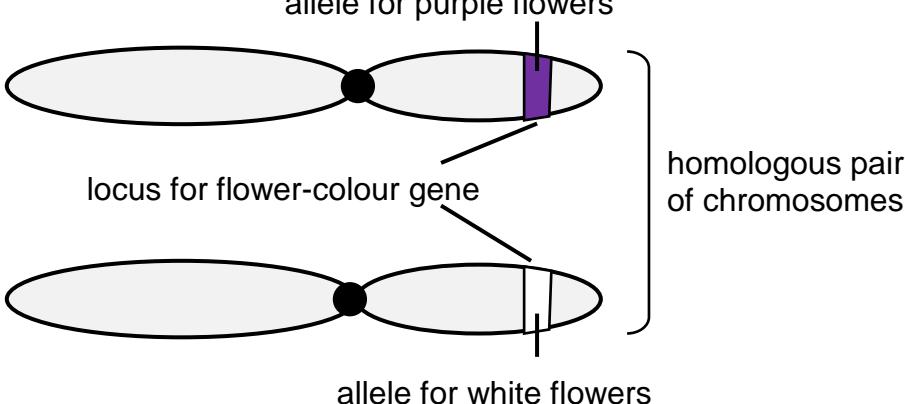
Table 1 below provides further definitions and explanations of key terms that you must know very well.

Table 1: Terms, their explanation and supporting diagrams and notes.

gene	a segment of DNA in a chromosome that contains the code for a particular characteristic	A diagram illustrating the relationship between a chromosome and its constituent DNA. On the left, a chromosome is shown as a pair of sister chromatids, each consisting of two vertical bars representing DNA segments. A bracket on the right side of the diagram groups these two chromatids together and is labeled 'DNA'. A line points from the word 'gene' in the adjacent text box to the central region of the chromosome, indicating that a gene is a segment of DNA located on a chromosome.
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alleles	different forms of a gene which occur at the same locus on homologous chromosomes	dominant allele (T) – tall plant recessive allele (t) – short plant
genotype	genetic composition of an organism	 <ul style="list-style-type: none"> • homozygous dominant (both alleles are dominant) • genotype TT • phenotype – tall
phenotype	the physical appearance of an organism based on the genotype, e.g. tall, short	 <ul style="list-style-type: none"> • homozygous recessive (both alleles are recessive) • genotype tt • phenotype – short
dominant allele	an allele that is expressed (shown) in the phenotype when found in the heterozygous (Tt) and homozygous (TT) condition	 <ul style="list-style-type: none"> • heterozygous (one dominant and one recessive allele) • genotype Tt • phenotype - tall
recessive allele	an allele that is masked (not shown) in the phenotype when found in the heterozygous (Tt) condition; only expressed in the homozygous (tt) condition	
heterozygous	two different alleles for a particular characteristic, e.g. Tt	
homozygous	two identical alleles for a particular characteristic, e.g. TT or tt	
monohybrid cross	only one characteristic or trait is shown in the genetic cross	Flower colour only, e.g. yellow or white flower – OR – shape of seeds only, e.g. round seeds or wrinkled seeds
dihybrid cross	two different characteristics shown in genetic cross	Example: flower colour, e.g. yellow or white flower – AND – shape of seeds, e.g. round seeds or wrinkled seeds

complete dominance	a genetic cross where the dominant allele masks the expression of a recessive allele in the heterozygous condition	- the allele for tall (T) is dominant over the allele for short (t) - offspring will be tall because the dominant allele (T) masks the expression of the recessive allele (t)	Tall (TT) x short (tt)  Tall (Tt)
incomplete dominance	cross between two phenotypically different parents produces offspring different from both parents but with an intermediate phenotype	a red-flowered plant is crossed with a white-flowered plant – with incomplete dominance, offspring will have pink flowers - intermediate colour.	red x white flower  pink flowers
co-dominance	cross in which both alleles are expressed equally in the phenotype.	red-flowered plant is crossed with a white-flowered plant – with co-dominance, the offspring has flowers with red and white patches	red x white flower  flowers with red and white patches
multiple alleles	more than two alternative forms of a gene at the same locus	blood groups are controlled by three alleles, namely I ^A , I ^B and i. all three alleles are present in a population, but an individual can only have two alleles	
sex-linked characteristics	traits that are carried in the sex chromosomes	Some examples: haemophilia and colour-blindness - alleles for haemophilia (or colour-blindness) are indicated as superscripts on the sex chromosomes, e.g. X ^H X ^H (normal female), X ^H X ^h (normal female), X ^h X ^h (female with haemophilia), X ^H Y (normal male), X ^h Y (male with haemophilia). Both haemophilia and colour blindness are caused by recessive alleles.	

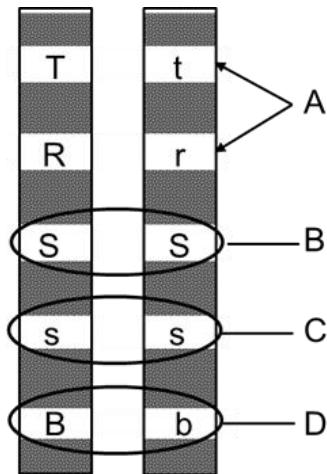
karyotype	the number, shape and arrangement of the chromosomes in the nucleus of a somatic cell	 XX / XY 23
cloning	process by which genetically identical organisms are formed using biotechnology	Dolly the sheep was cloned using a diploid cell from one parent; therefore, it had the identical genetic material of that parent
genetic modification	manipulation of the genetic material of an organism to get desired changes	An example: insertion of the human insulin gene into the plasmid of bacteria so that the bacteria produce human insulin
human genome	mapping of the exact position of all the genes in all the chromosomes of a human	Example: haemophilia is the last gene on the X chromosome; gene number 3 on chromosome 4 is responsible for a particular characteristic
homologous pair of chromosomes	<p>a set of one maternal and one paternal chromosome that pair up with each other inside a cell during meiosis – homologous chromosomes are the same size and shape, and carry the same or similar alleles</p> 	

Activity 1: Traits, genes and alleles

1. The paired letters in the diagram below represent alleles of a gene. A number of genes for different characteristics are shown. Write down the relevant letter (A – D) for:

- a) The homozygous dominant state
- b) Two alleles from different genes
- c) The homozygous recessive state
- d) The heterozygous state

(4)



2. What is the relationship between a gene and a protein? (2)
3. What is an allele? (2)
4. What terms describe a pair of alleles that are:
 - a) the same?
 - b) different? (2)
5. Write a definition of homologous chromosomes using the terms “genes” and “alleles”. (3)
6. How are alleles represented? (1)

7. Fill in the table below with the missing genotype, phenotype (dominant or recessive), or alleles (TT, Tt, tt) (6)

Genotype	Phenotype	Alleles
homozygous dominant		
	short	t / t

8. Draw a pair of homologous chromosomes. Label the chromosomes with two sets of genes, one with homozygous dominant alleles, one with homozygous recessive alleles and one with heterozygous alleles. (5)
(25)

Mendel as father of genetics

Gregor Mendel, an Austrian monk (a type of priest), is regarded as the father of genetics for his work on garden pea plants that helped explain **how genes are passed from parents to offspring**.

Mendel's work on the genetics of peas began with the observation of peas to determine what traits were inherited. He noticed at least 7 traits that appeared to be inherited.

These traits were (see Table 2):

- seed shape
- seed colour
- pod shape (pod – the container holding a plant's seeds)
- pod colour
- flower colour
- flower position (whether axial – on the side, or terminal – on top)
- stem length



Figure 1: Gregor Mendel

Table 2: Traits compared by Mendel

Seed		Pod		Flower		Size
shape	colour	shape	colour	position	colour	
round	yellow	full	green	axial	purple	tall
wrinkled	green	constricted	yellow	terminal	white	short

Mendel's Experiments

- Mendel noticed that garden peas occur in at least two heights – tall (T) and short (t)
- Since peas are self-pollinating, tall peas tend to produce tall peas, and short peas produce short peas.
- Mendel's first genetic cross involved tall peas cross-pollinated with short peas
- The result: in the first group of offspring (the **F₁** generation), the cross yielded only tall peas
- Mendel then took the **F₁** peas and crossed them with themselves (interbreeding) to produce a second group of offspring (an **F₂** generation)
- The result: tall and short peas, in a ratio of 3:1 (3 tall: 1 short or dwarf)

This experiment of Mendel's may be illustrated in the diagram as in Figure 2 below.

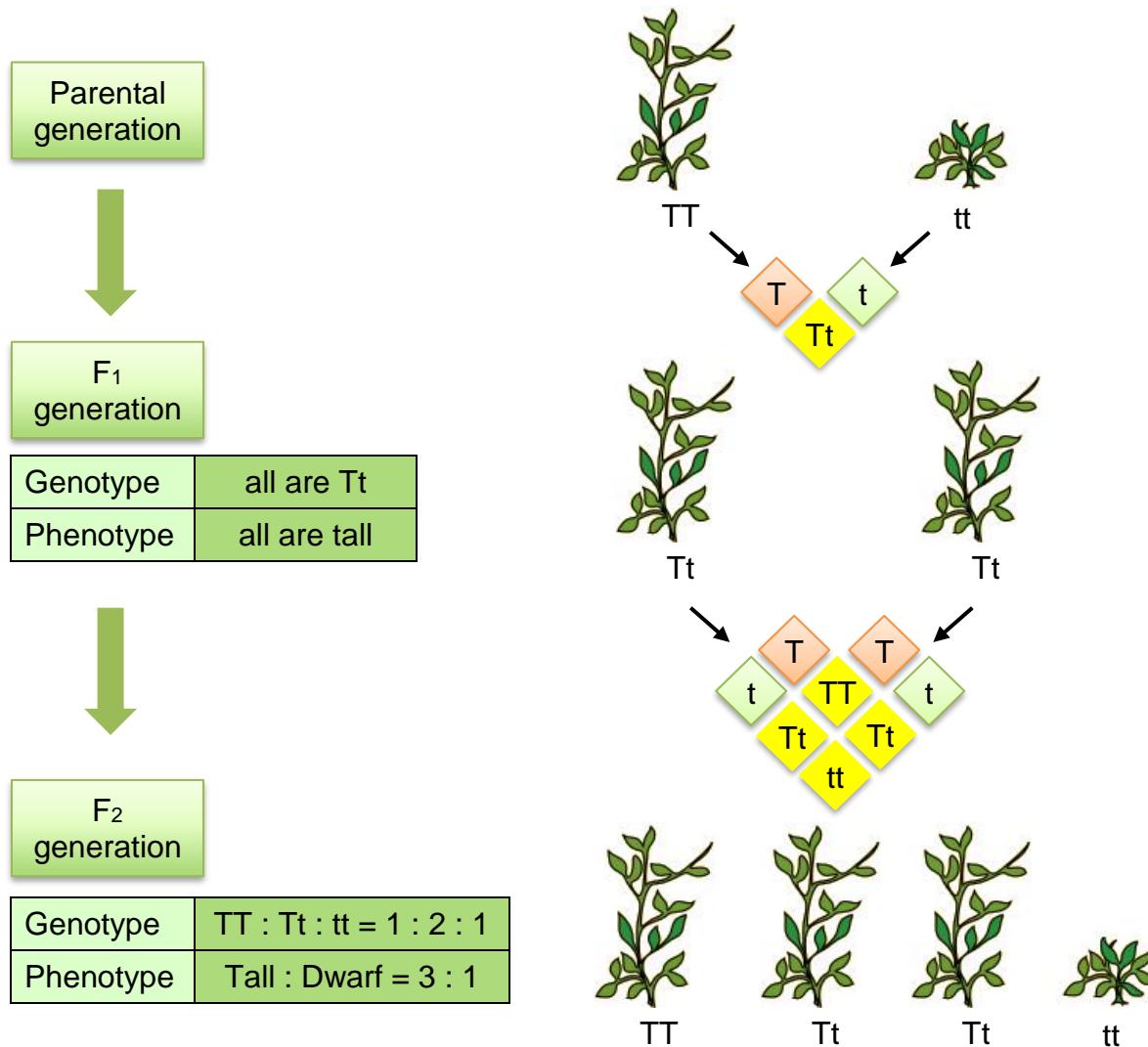


Figure 2: A genetic cross between a tall pea plant and a short (dwarf) plant

Mendel's Laws of Inheritance

Mendel formulated the following laws from his experiments.

Mendel's First Law of Inheritance: Law (principle) of Segregation

- Each trait is controlled by two factors (now known as alleles) situated on homologous chromosomes.
- When gametes form during meiosis, the two factors (alleles) are separated or segregated. A gamete contains one of the two factors (alleles) from each parent.

Mendel's Second Law of Inheritance: Law of Dominance

- Certain alleles of a gene exist in either a dominant or a recessive form.
- If the pair of alleles are different (one dominant, one recessive), the phenotype will only show the dominant trait.

Mendel's Third Law of Inheritance: Law (principle) of Independent Assortment

- Due to random arrangement of chromosomes at the equator during meiosis (gamete formation), any one of the two alleles of ONE characteristic can sort with any one of ANOTHER characteristic.
- The alleles of different genes move independently of each other into the gametes. They can therefore appear in the gametes in different combinations.

Genetic diagrams

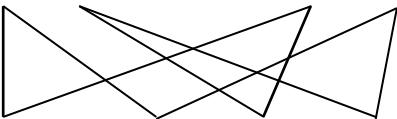
- It is important to start with a **PAIR** of alleles in the mother and another **PAIR** in the father because each individual inherits TWO alleles for a gene – one maternal and one paternal (the bivalent chromosomes).
- This **pair** may be identical (HOMOZYGOUS) or different (HETEROZYGOUS) alleles.
- Alleles are represented by **letters**: CAPITALS (for dominant alleles) or small letters (for recessive alleles).
- P stands for the **Parent** generation

- F stands for the offspring (**Filial generation**) – remember **F** for Family.

F_1 generation is the first filial generation, F_2 the second.

Genetic crosses

It is important to learn the **exact** layout because marks are allocated for the layout as well as the correct working out of the cross. Use the following genetic diagram or template to solve all genetic crosses.

Layout of a genetic diagram			Explanation												
P₁	Phenotype	x	✓												
Meiosis	Genotype	x	✓												
	Gametes	x	✓												
Fertilisation															
F₁	Genotype		✓												
	Phenotype		✓												
P ₁ and F ₁ ✓															
Meiosis and fertilisation ✓															
OR															
P₁	Phenotype	x	✓												
Meiosis	Genotype	x	✓												
	Gametes	x	✓												
Fertilisation	<table border="1" style="margin-left: auto; margin-right: auto;"> <tr> <td>Gametes</td><td></td><td></td></tr> <tr><td></td><td></td><td></td></tr> <tr><td></td><td></td><td></td></tr> <tr><td></td><td></td><td></td></tr> </table>			Gametes											
Gametes															
F₁	Phenotype		✓												
	Genotype		✓												
P ₁ and F ₁ ✓															
Meiosis and fertilisation ✓															

IMPORTANT: You may also be asked to work out the ratio or % chance of the various pheno-/genotypes occurring. So, if there are 4 possible geno- or phenotypes in total and only 1 having a particular phenotype, it will be a 1 in 4 ratio (25% chance)

You need to read genetics questions with great care to find the clues concerning:

- which characteristic (phenotype) is being examined, e.g.: shape of seeds
- which allele of the pair is dominant, e.g.: round
- whether the parents are homozygous or heterozygous for the characteristic: e.g.: both are heterozygous
- what letters are to be used (are they given OR must you choose letters), e.g.: R (dominant allele) and r (recessive allele)
- show how the alleles are separated during meiosis to form gametes
- show all the possible ways the sperm and egg can combine during fertilisation
- distinguish the phenotypes from the genotypes

Monohybrid crosses

Monohybrid crosses refer to genetic crosses that involve only a single characteristic or trait. **Dihybrid** crosses involve two characteristics or traits.

The following monohybrid crosses are dealt with:

- monohybrid crosses with complete dominance
- monohybrid crosses with incomplete dominance
- monohybrid crosses with co-dominance
- sex determination
- inheritance of sex-linked diseases
- multiple alleles e.g.: blood grouping

Monohybrid crosses with complete dominance

Mendel's work as discussed above shows monohybrid crosses with complete dominance. In complete dominance, the dominant allele masks or blocks the expression of the recessive allele in the heterozygous condition.

The following example represents a genetic cross which shows complete dominance.

Genetic problem 1

Seeds can be round or wrinkled. Use a genetic cross to show the genotype and the phenotype of the F₁ generation when two heterozygous plants with round seeds are crossed. The allele for round seeds is dominant over the allele for wrinkled seed. Use the letter R for round and r for wrinkled seeds.

P₁	Phenotype	Round seeds	x	Round seeds									
	Genotype	Rr	x	Rr									
Meiosis	Gametes	R	r	R									
			x										
			R	r									
Fertilisation													
F₁	Genotype	RR	Rr	Rr									
	Phenotype	round seeds	round seeds	round seeds									
				wrinkled seeds									
				OR									
P₁	Phenotype	round seeds	x	round seeds									
	Genotype	Rr	x	Rr									
Meiosis	Gametes	R	r	R									
			x										
			R	r									
Fertilisation													
				<table border="1" style="border-collapse: collapse; width: 100%; text-align: center;"> <tr> <td style="padding: 2px;">Gametes</td> <td style="padding: 2px;">R</td> <td style="padding: 2px;">r</td> </tr> <tr> <td style="padding: 2px;">R</td> <td style="padding: 2px;">RR</td> <td style="padding: 2px;">Rr</td> </tr> <tr> <td style="padding: 2px;">r</td> <td style="padding: 2px;">Rr</td> <td style="padding: 2px;">rr</td> </tr> </table>	Gametes	R	r	R	RR	Rr	r	Rr	rr
Gametes	R	r											
R	RR	Rr											
r	Rr	rr											
F₁	Phenotype	round seeds	round seeds	round seeds									
				wrinkled seeds									
	Genotype	RR	Rr	Rr									
				rr									

Genotypic Ratio : 1RR : 2Rr : 1rr (1:2:1)

Phenotypic Ratio: 3 Round seeds: 1 Wrinkled seed (3:1)

- A cross between two heterozygous (Rr) parents produces 25% homozygous dominant (RR), 50% heterozygous (Rr) and 25% homozygous recessive (rr) offspring. As such, the phenotypic ratio is 3 dominant : 1 recessive.

Activity 2: Monohybrid crosses with complete dominance

1. Mendel crossed homozygous plants with green seeds with homozygous plants which had yellow seeds and found that all the offspring were yellow (Figure 3). Construct a genetic diagram (using the above template) to show the F_1 and F_2 generations.

Choose a letter for each allele. Note: use one letter: capitalised for the dominant allele and small (lower-case) for the recessive allele. (6 + 6)

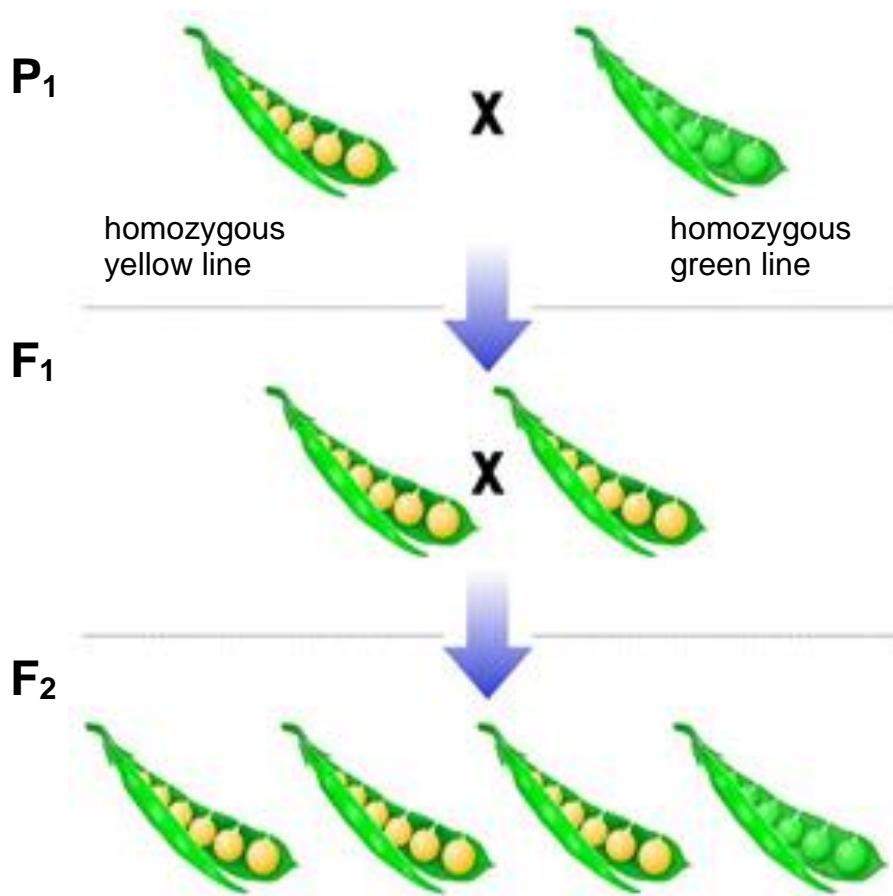


Figure 3: Monohybrid cross showing inheritance of seed colour

2. The ability to roll one's tongue (Figures 4 and 5) is a dominant characteristic. Explain (without a genetic diagram) why it is possible for a non-roller child to be born to parents who can both roll their tongues. Use the letters R and r in your answer. (3)

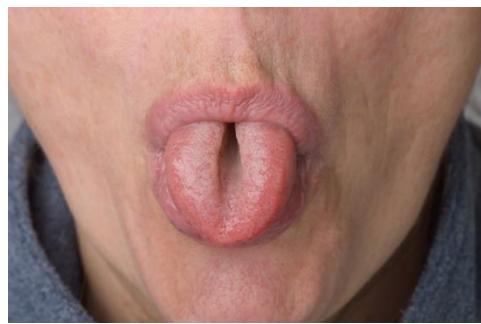


Figure 4: Tongue-roller



Figure 5: Non-roller

(15)

Monohybrid cross with incomplete dominance

Incomplete dominance is a cross between two phenotypically different parents where no allele of the gene is either dominant or recessive. The offspring is different to both parents and the alleles blend to form a **new phenotype**.

The following problem represents a genetic cross which shows incomplete dominance:

Genetic problem 2

A homozygous red-flowering plant crossed with a homozygous white-flowering plant will produce plants that have pink flowers (Figure 6).

Complete a genetic diagram to show how this is possible using the letter **R** for red and **W** for white.

P₁	Phenotype	Red	x	White
	Genotype	RR	x	WW
Meiosis				
	Gametes	R	R	x
				W W
Fertilisation				
	Gametes	R	R	
	W	RW	RW	
	W	RW	RW	
F₁	Genotype	RW	RW	RW RW
	Phenotype	All Pink		

The characteristic being investigated is flower colour; the alleles are red (**R**) and white (**W**).

The genotype of the parents will be **RR** and **WW** as they are homozygous where both alleles for the gene are the same.

The fact that the offspring have a new phenotype **pink** which is formed from red and white, tells you that there is no dominant or recessive allele.

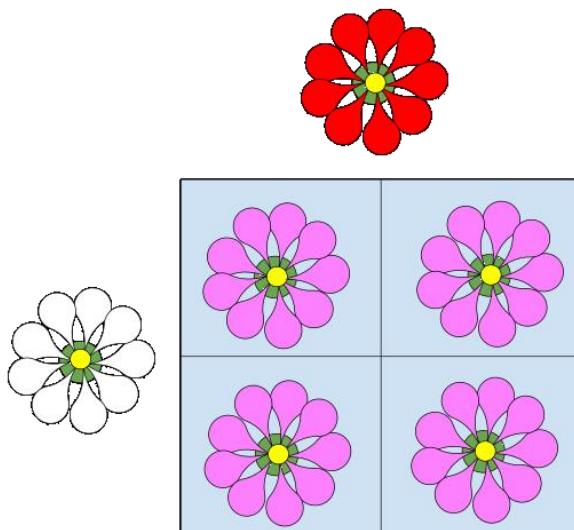


Figure 6: Monohybrid cross with incomplete dominance

Activity 3: Monohybrid cross with incomplete dominance

1. Two grey mice were mated. Some of the offspring were grey, others black and some white. How is this possible? Do a genetic cross to explain this result. (6)
2. Incomplete dominance is seen in the inheritance of hypercholesterolemia (high blood cholesterol levels). **H** represents the allele for very high levels and **L** for low levels.
 - 2.1 Sipho and Andiswa are both heterozygous for this characteristic and both have high cholesterol levels but not as high as their daughter Sihle who has levels that are six times above normal. She is homozygous for high cholesterol levels. Do a full genetic diagram to explain your answer. (7)
 - 2.2 What is the percentage chance that their next child will have ...
 - a) low cholesterol levels? (1)
 - b) extremely high cholesterol levels like Sihle? (1)(15)

Monohybrid crosses with co-dominance

In co-dominance **both** alleles of the gene are equally dominant so both will be expressed equally in the phenotype of the offspring.

The following problem represents a genetic cross which shows co-dominance:

Genetic problem 3

In cattle three colour variations are possible (Figure 7): red, white or red and white in patches. This comes about due to a red allele (**R**) and a white allele (**W**) for coat colour. Do a genetic cross between a red bull and a white female to show the possible offspring.

P₁	Phenotype	Red coat	x	White coat									
	Genotype	RR	x	WW									
Meiosis		R R	x	W W									
	Gametes												
Fertilisation		<table border="1"><tr><td>Gametes</td><td>R</td><td>R</td></tr><tr><td>W</td><td>RW</td><td>RW</td></tr><tr><td>W</td><td>RW</td><td>RW</td></tr></table>	Gametes	R	R	W	RW	RW	W	RW	RW		
Gametes	R	R											
W	RW	RW											
W	RW	RW											
F₁	Genotype	RW	RW	RW									
	Phenotype	All red with white patches											



Figure 7: Co-dominance in cattle showing 3 coat colours

The characteristic being investigated is coat colour which is controlled by two alleles **R** and **W**. The **F₁** offspring show both phenotypes together so this is co-dominance.

The offspring will have a genotype of **RW** which is red and white patches.

Do this cross for yourself, and then cross two of the F₁ offspring to see what the possibilities would be in the F₂ generation.

Note: Inheritance of blood groups is also partly an example of co-dominance when the alleles I^A and I^B are involved. This will be dealt with later.

Sex determination

The following problem represents a genetic cross which shows inheritance of sex

Genetic problem 4:

A couple has three sons and the woman is pregnant again. Show by means of a genetic cross what the percentage chance is of the couple having a baby girl

P ₁	Phenotype	Male	x	Female
	Genotype	XY	x	XX
Meiosis				
	Gametes	X	Y	x
Fertilisation				
		Gametes	X	Y
		X	XX	XY
		X	XX	XY
F ₁	Genotype	XX	XX	XY XY
		2 (XX)		2(XY)
	Phenotype	Female (50%)		Male (50%)
	Phenotypic ratio	1:1		

The genetic cross above shows that the percentage chance of having a boy or a girl is 50%.

In humans, there are 46 chromosomes (i.e. 23 from the mother and 23 from the father). Of these 46 chromosomes, 44 control the appearance, structure and functioning of the body. These are called **autosomes**. The remaining pair determines the sex of the individual and are called the **gonosomes**. In a female the gonosomes are two large X chromosomes and in the male there is one large X chromosome and a smaller Y chromosome.

Each species has its own unique number, shape and size of chromosomes – this is called the **karyotype**.

Examine the two karyotypes below to see if you can identify which comes from a female and which from a male.

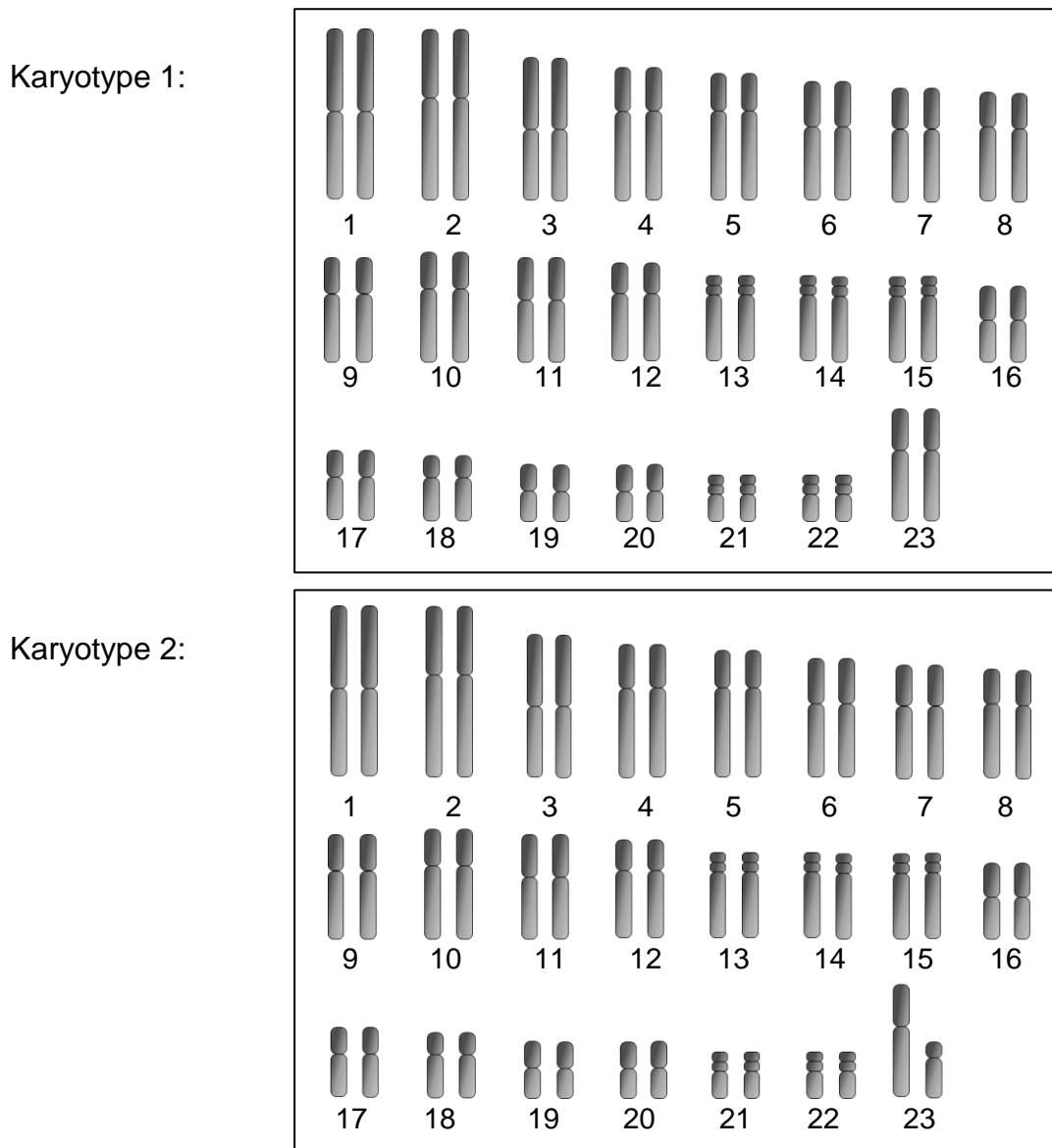


Figure 8: Female and male karyotypes

After meiosis, an egg cell will have 22 autosomes + an X gonosome. A female will have two large X gonosomes (as in karyotype 1 – see pair 23), while a male will have an X gonosome and a Y gonosome (as in karyotype 2 – see pair 23).

Males thus have two types of sperm: half will have 22 + X chromosomes, and the other half will have 22 + Y chromosomes. Depending on which sperm reaches the egg, there is a 50% chance of the zygote being male and a 50% chance of the zygote being female.

Sex-linked inheritance

Although most of the bodily characteristics are carried on the 22 pairs of autosomes, there are a few characteristics carried on the gonosomes only. Thus, for example, the gene for hair growing on the inside of the pinna (Figure 9) is carried on the Y chromosome, so only men will have this characteristic.

Certain sex-linked genetic disorders are carried on the allele found on the X chromosome only. Two of these disorders are colour blindness and haemophilia.

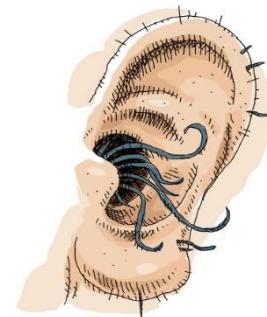


Figure 9: Hairy

- A person is **colour-blind** if unable to tell different colours apart. For example, red-green colour-blindness is caused by an absence of the proteins that make up the red or green cones (photoreceptors) in the retina of the eye resulting in the person not being able to tell the difference between red and green.
- **Haemophilia** is the inability of the blood to clot due to lack of a blood clotting factor. If the sufferer were to cut themselves, the wound would continue to bleed until a clotting factor is transfused in hospital.
- Colour-blindness and haemophilia is caused by the recessive allele on the X-chromosome normally shown as (X^b) for colour-blindness and (X^h) for haemophilia
- As a result, men who have only one X chromosome, have a greater risk of inheriting these disorders.
- Women, on the other hand, have a much lower chance of inheriting two X chromosomes which both carry the recessive allele for the disorder. If a woman inherits one X chromosome with the recessive allele for the disorder, she is called a **carrier** as she does not show signs of the disorder but can pass it on to her children.

Tables 3 and 4 below relate the inheritance of haemophilia and colour-blindness.

Table 3: Inheritance of haemophilia

Genotype	Phenotype
$X^H X^H$	Normal female
$X^H X^h$	Normal female (carrier)
$X^h X^h$	Haemophiliac female
$X^H Y$	Normal male
$X^h Y$	Haemophiliac male

Table 4: Inheritance of colour-blindness

Genotype	Phenotype
$X^B X^B$	Female with Normal vision
$X^B X^b$	Normal Female (carrier)
$X^b X^b$	Colour-blind female
$X^B Y$	Normal male
$X^b Y$	Colour-blind male

Do not add any letter to the Y chromosome since the Y chromosome does not have an allele to counteract the recessive allele for haemophilia and colour-blindness.

Activity 4: Sex-linked diseases

1. Haemophilia is a sex-linked disease caused by the presence of a recessive allele (X^h).
A normal father and heterozygous mother have children. Construct a genetic cross to determine the possible genotype and phenotype of the children of the parents. (6)
2. Explain why the chances of men having a sex-linked disorder is much higher than it is for women. (4)

In an exam you may be asked to do other sex-linked disorders other than haemophilia and colour-blindness. Unless stated otherwise, follow the same format as in haemophilia and colour-blindness.

3. Read the following extract on cystic fibrosis and answer the questions that follow.

Cystic Fibrosis (CF)

CF is a progressive, genetic disorder caused by a recessive allele on chromosome number 7. One in twenty people of European descent carry the CF allele. One in 400 couples of European descent will be carriers of CF.

The disorder causes persistent lung infections and limits the ability to breathe over time. In people with cystic fibrosis, the defective gene causes a thick, build-up of mucus in the lungs and it clogs the airways and traps bacteria leading to infections and then eventually respiratory failure.

- 3.1 Explain why cystic fibrosis is not a sex-linked disease. (2)
- 3.2 Use a genetic cross to show what percentage of children will be affected if one of the parents is heterozygous and the other is homozygous normal. The recessive allele is represented as **b**. (6)
- 3.3 Maggie and William want to start a family, but Maggie's brother had cystic fibrosis. She doesn't want her own child to suffer as her brother did. Maggie and William decided to visit a genetic counsellor. Explain how this may help Maggie and William in their decision. (2)
(20)

Multiple alleles – blood type / group

The genetic crosses dealt with thus far involved two alleles of a gene, e.g.: T or t, R or W. Sometimes a characteristic is however controlled by more than two alleles. Blood type (or blood grouping) is an example of such a characteristic.

There are four blood types in humans: A, B, AB or O. These **phenotypes** are controlled by three alleles but each person still inherits two alleles. It is very important to know how to name (or write down) these three alleles as they are very specific to blood groups namely **I^A**, **I^B** or **i**.

I^A is co-dominant to **I^B** whereas **i** is recessive to both. The genotypes for each blood group can then be specified as follows:

Genotype	Blood group
I ^A I ^A	A
I ^A i	A
I ^B I ^B	B
I ^B i	B
I ^A I ^B	AB
ii	O

Use of blood groups in paternity testing

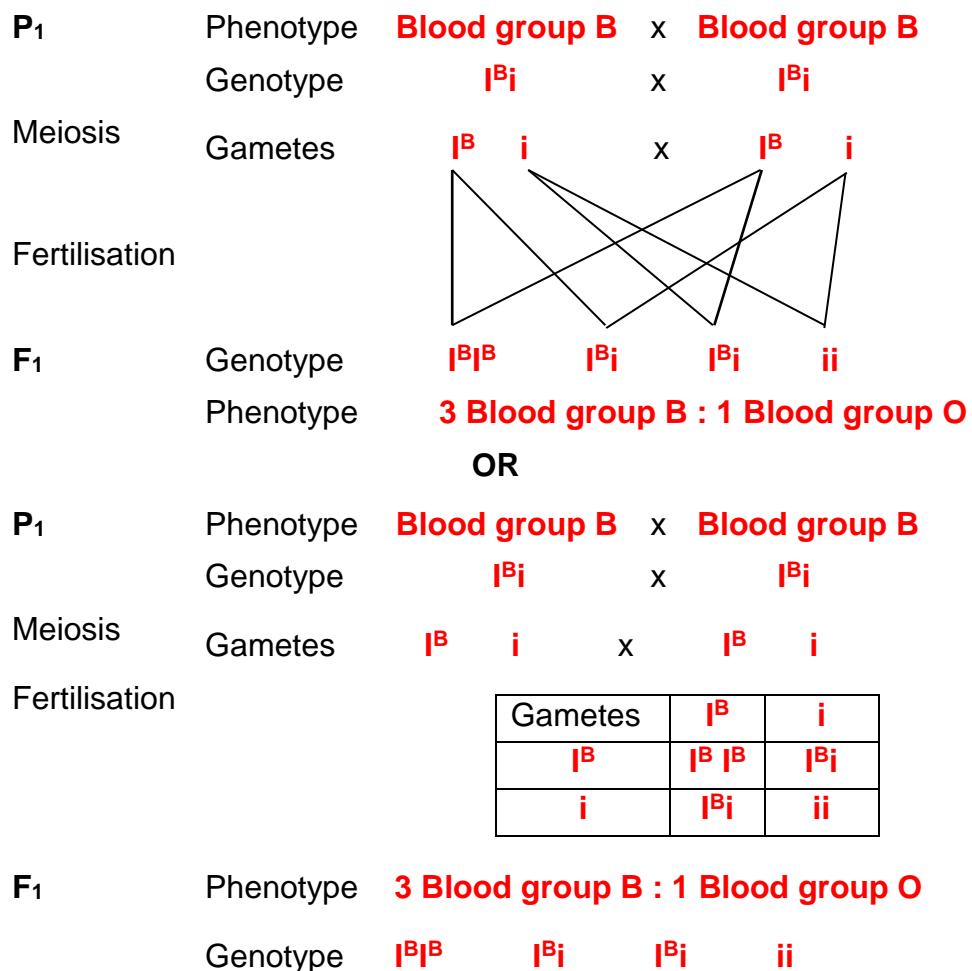
The blood groups of the mother, possible father and child must be compared. If the blood groups of the adults do not correspond to or match the child's blood group then this man is not the father. If the blood groups of the adults correspond to or match the child's blood group, then there is a possibility that the man is the father and other tests need to be done as other men may have the same blood group.

Only DNA profiling can be conclusive as it looks at the similarities between the nucleotides in the DNA of the father and the child. Each DNA profile is unique to an

individual. 50 % of the DNA fragments / bands / bars are derived from the mother and 50 % from the father. If 50 % of the DNA fragments / bands / bars correspond with the father, then it can be claimed that he is the father of the child. DNA is viewed as more reliable evidence of paternity than the use of blood groups.

Example of a monohybrid cross using blood types:

A man and a woman both have blood group **B**. Use a genetic cross to show how it is possible for them to have a child with blood group **O**.



Activity 5: Monohybrid crosses using blood types

1. If the child has blood group **O** and the mother blood group **A**, could the man with blood group **AB** be the father of that child? Use a genetic diagram to explain your answer. (6)
2. Human blood groups are controlled by multiple alleles.
 - a) List all the alleles that control human blood groups. (3)
 - b) How many of the alleles named in a) can any individual inherit? (1)

- c) Give a reason for your answer to question b). (1)
 - d) Which 2 alleles are co-dominant in the inheritance of blood groups? (2)
 - e) A man has blood group **A** and his wife blood group **B**. Their first child has blood group **AB** and the second child blood group **O**. What can one conclude about the blood groups of their future children? (3)
- (15)

Dihybrid crosses

Dihybrid crosses involve **two pairs** of alleles representing **two different** characteristics, e.g.: the height of a plant and the colour of its seeds.

According to the Law of Independent Assortment, alleles of different genes move (segregate) independently of each other into the gamete. They therefore appear on the gametes in different combinations.

Work through the following example of a dihybrid cross, and remember that the alleles for each characteristic could be either homozygous or heterozygous.

Example of a dihybrid cross

In pea plants, the allele for tallness (**T**) is dominant and the allele for shortness (**t**) is recessive. The allele for purple flowers is dominant (**P**) and the allele for white flowers is recessive (**p**). Two plants, heterozygous for both tallness and purple flowers were crossed.

- **Step 1:** Decide whether this concerns a monohybrid or a dihybrid cross.

*Since two characteristics of each plant are mentioned (phenotypes: **height of plant + colour of flower**), it must be a dihybrid cross.*

- **Step 2:** Choose/ use letters to represent the alleles for the gene responsible for each characteristic.

Let T = the allele for tall plants

Let t = the allele for short plants

Let P = the allele for purple flowers

Let p = the allele for white flowers

- **Step 3:** Write down the phenotype of the two parents that would be producing gametes.

tall purple X tall purple (as per question)

- **Step 4:** Write down the genotype of the parents.

TtPp X TtPp

- **Step 5:** Show the gametes that each parent produces after meiosis. Each gamete must have two letters (dihybrid) – one from each characteristic.

N.B. Remember Mendel's Law of Independent Assortment.



- **Step 6:** Draw and complete a punnet square by writing in the combination of alleles in each block.

P₁	Phenotype	Tall, purple	x	Tall, purple
	Genotype	TtPp	x	TtPp
Meiosis	Gametes	TP Tp tP tp	x	TP Tp tP tp

Fertilisation	Gametes	TP	Tp	tP	tp
	TP	TTPP	TTPp	TtPP	TtPp
	Tp	TTPp	TTpp	TtPp	Ttpp
	tP	TtPP	TtPp	ttPP	ttPp
	tp	TtPp	Ttpp	ttPp	ttpp

F₁	Genotype	9 different genotypes, as in the table above
	Phenotype	9 tall, purple flowered plants
		3 short, purple flowered plants
		3 tall, white flowered plants
		1 short, white flowered plant

- **Step 7:** Determine the phenotypic ratios from the genotypes in the punnet square

Phenotypic ratio: 9:3:3:1

If there is one capital letter for the allele in the **F₁** generation, then that trait (characteristic) shows in the phenotype; if there are small letters then the recessive trait shows.

Activity 6: Dihybrid crosses

- Two characteristics of an animal (length of the ears and shape of the lip) were studied. Each of these characteristics has two variations: Ears may be long or short, and the lip may be a wide or pointed.

A male animal homozygous for wide lips (**LL**) and heterozygous for short ears (**Ee**) is crossed with a female animal that is heterozygous for wide lips (**Ll**) and homozygous for long ears (**ee**).

- What term describes a genetic cross involving two characteristics? (1)
 - Give the
 - dominant phenotype for the length of ears (1)
 - recessive phenotype for the shape of the lip (1)
 - possible genotype/s for an animal with short ears and a pointed lip (1)
 - A male animal with genotype **EELl** is crossed with a female animal with genotype **Eell**. List all the possible gametes that could be produced by the male animal. (2)
 - Explain how Mendel's Law of Independent Assortment applies to parents with **LlEe** genotypes during gamete formation. (4)
2. In humans the allele for short fingers (brachydactyly – a shortening of the fingers and toes), represented by **B**, is dominant over the allele for normal fingers (**b**). The allele for curly hair (**H**) is dominant over the allele for straight hair (**h**).

Andrew, with genotype **Bbhh**, married Susan, with genotype **bbHh**.

- How do Andrew and Susan's phenotypes differ from each other? (2)
 - List all possible genotypes of the gametes produced by Andrew. (2)
- (14)

Genetic lineage (Pedigree diagrams)

A pedigree diagram (also called a family tree) is used to study the inheritance of characteristics in a family over a number of generations.

The pedigree diagram in Figure 10 (see next page) shows inheritance of eye colour in humans over three generations of a family. Brown eye colour (**B**) is dominant over blue eye colour (**b**).

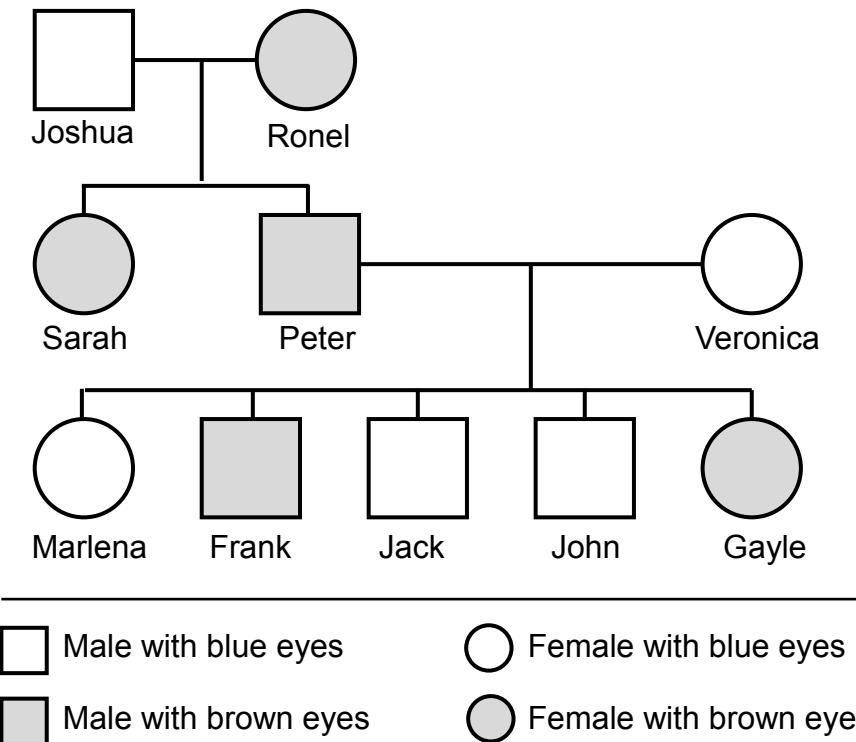


Figure 10: Pedigree diagram

- Squares represent males and circles represent females
- The horizontal line between a square (Joshua) and a circle (Ronel) shows that they have mated.
- The vertical line flowing from the horizontal line represents the offspring (Sarah and Peter) of the two parents (Joshua and Ronel).

Remember the following steps when interpreting pedigree diagrams:

- Step 1:** Study any key and opening statement/s and look for dominant and recessive characteristics and phenotypes.
Brown eye colour (B) is dominant over blue eye colour (b) – as stated in the problem
- Step 2:** Write in the phenotypes of all the individuals as given in the problem.
 - Joshua, Jack and John are males with blue eyes.*
 - Veronica and Marlena are females with blue eyes.*
 - Peter and Frank are males with brown eyes.*
 - Ronel, Sarah and Gayle are females with brown eyes.*
- Step 3:** Fill in the genotype of all the individuals with the recessive condition – it must have two recessive alleles (two lower case letters, e.g. **bb**).

*Joshua, Veronica, Marlena, Jack and John will have the genotype 'bb'.
The recessive characteristic only shows up in the homozygous condition*

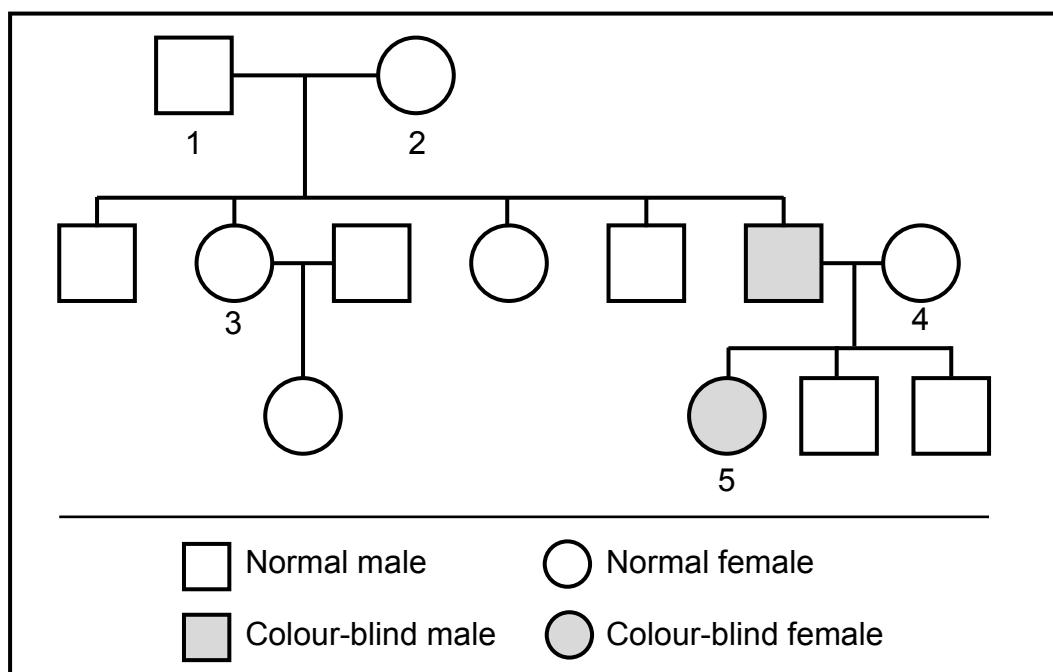
- **Step 4:** For every individual in the diagram that has the recessive condition, it means that each allele was obtained from each of the parents. Work backwards and fill in one recessive allele for each parent.
- **Step 5:** If the parents showed the dominant characteristic, fill in the second letter which represents the dominant allele (a capital letter, e.g. **B**).

The genotype of Peter is 'Bb' – working backwards from the offspring Marlena or Jack or John who are homozygous recessive. This means that one of the recessive alleles of Marlena, Jack and John, i.e. 'b', must have come from parent Peter and the other one from parent Veronica

- **Step 6:** Any other individual showing the dominant characteristic will most likely be homozygous dominant (**BB**) or heterozygous dominant (**Bb**).
Ronel could be homozygous dominant (BB) or heterozygous dominant (Bb)

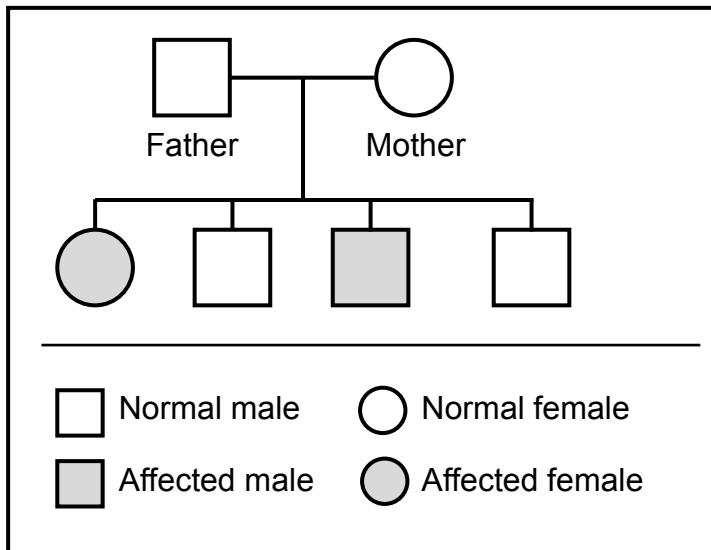
Activity 7: Pedigrees

1. The pedigree diagram below shows the inheritance of colour-blindness (also called Daltonism) in a family. Colour-blindness is sex-linked and is caused by a recessive allele (**d**). The ability to see colour normally is caused by a dominant allele (**D**).

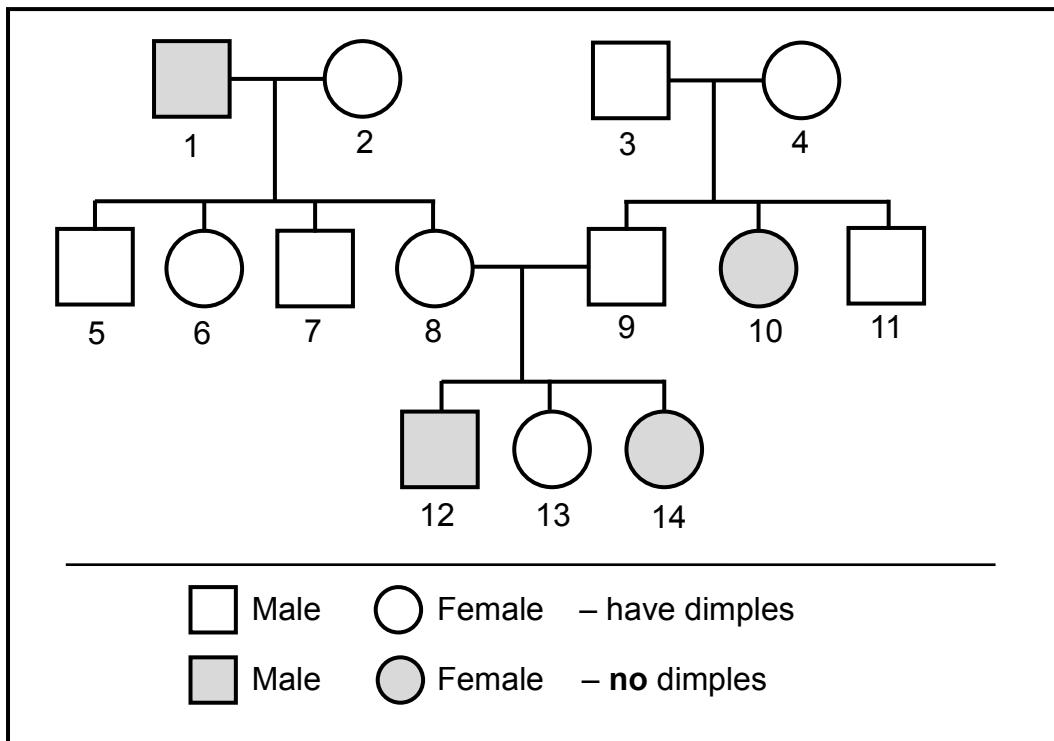


Inheritance of colour-blindness

- 1.1 How many of the male offspring of parents 1 and 2 were normal? (1)
- 1.2 What percentage of males in this pedigree diagram are affected? Show your workings. (2)
- 1.3 State the genotype of
- Individual 2 (1)
 - Individual 5 (1)
- 1.4 If individual 5 marries a normal male, what percentage of their daughters will have an allele for colour-blindness, but will not be colour-blind? (1)
2. The pedigree diagram below shows the pattern of inheritance of a certain genetic disorder controlled by a recessive allele. The dominant allele is represented by **N** and the recessive allele by **n**.
- Explain why both parents must be heterozygous for this characteristic. (2)
 - Give the possible genotype(s) of the normal children. (2)
 - Provide evidence from the pedigree diagram to show that this characteristic is not sex-linked. (3)



3. Use the pedigree diagram below to answer the questions about dimples (small depressions on the cheeks when smiling). The dimple allele (**D**) controls whether a person has dimples or does not have dimples. The allele for having dimples is dominant to the allele for not having dimples (**d**).



- 3.1 How many family members have dimples? (1)
- 3.2 What is the genotype of the individuals?
- 3 (1)
 - 4 (1)
- 3.3 State whether the following individuals are homozygous or heterozygous for having dimples:
- 2 (1)
 - 9 (1)
- 3.4 State the family relationship between individual 12 and individual 2. (1)
(19)

Mutations

A mutation is caused by a permanent change to the DNA of a cell. Mutations can be harmless, harmful or useful.

Harmless mutations mostly

- involve changes to the non-coding DNA (which makes up 98,5% of the DNA).
- This DNA is not involved in making proteins.

- It does not affect the structure or functioning of the cell/organism.
- Examples: freckles, blonde hair, baldness.

Harmful mutations

- change the DNA responsible for the production of a specific protein.
- This would cause changes to the organism's physical appearance or functioning due to an incorrect / defective protein being made.
- may cause a genetic disorder. Examples will be discussed below.

Useful mutations

- also change the DNA responsible for the production of a specific protein.
- If the protein made increases the organism's chance of survival, it would be seen as a useful mutation.
- If the gene is passed on, it will lead to genetic variation that is advantageous to the individual.
- Genetic variation is important to the processes of natural selection.
- In natural selection, organisms with traits that allow them to survive in the environment are more likely to pass on their genes.
- Natural selection (which will be discussed in chapter 9) is responsible for these mutations either being passed on to the future generations or not.

Mutations can occur in **genes** or **chromosomes**.

Gene mutations

Gene mutations occur during **replication** if a base pair is added, left out or doubled up. This changes the sequence of bases in DNA. Examples of gene mutations are haemophilia, colour-blindness, sickle cell anaemia, albinism.

- **Haemophilia** and **colour blindness** are sex-linked gene mutations on the **X**-chromosome.
- **Sickle cell anaemia** is an autosomal disease common in Central Africa, India and South America. It is caused by a gene mutation which results in a faulty haemoglobin molecule being formed. The red blood cells which are made have a half-moon shape (hence the term 'sickle'). Not only can these cells not carry enough oxygen (resulting in anaemia), but the shape means that the cells stick to each other blocking small capillaries. This causes damage in organs such as the brain and kidneys.

- **Albinism** is a rare group of genetic disorders which results in a lack of the pigment melanin. It is caused by a recessive gene mutation which prevents the normal development of colour in skin, hair or eyes.

Unfortunately, there are many prejudices towards albinos. They are often portrayed as villains in movies, are regarded as bringing bad luck, and are at times murdered for their body parts.

Albinos have weak eyes that are light sensitive and have a very light skin that is susceptible to skin cancer. They are perfectly normal in all other respects.

Chromosome aberrations

Chromosome aberrations occur during Anaphase I if the chromosomes of a bivalent do not separate. Both chromosomes go to the same pole, and thus the chromosome number of the gametes changes. An example of a chromosome aberration is Down syndrome.

Down Syndrome was discussed in Chapter 2 under abnormal meiosis. During Anaphase I / II, the non-disjunction of chromosome pair 21 will lead to the formation of a gamete with an extra (or one less) chromosome number 21. If this gamete fuses with a normal gamete, zygote with 3 (or only 1) chromosomes number 21 will form.

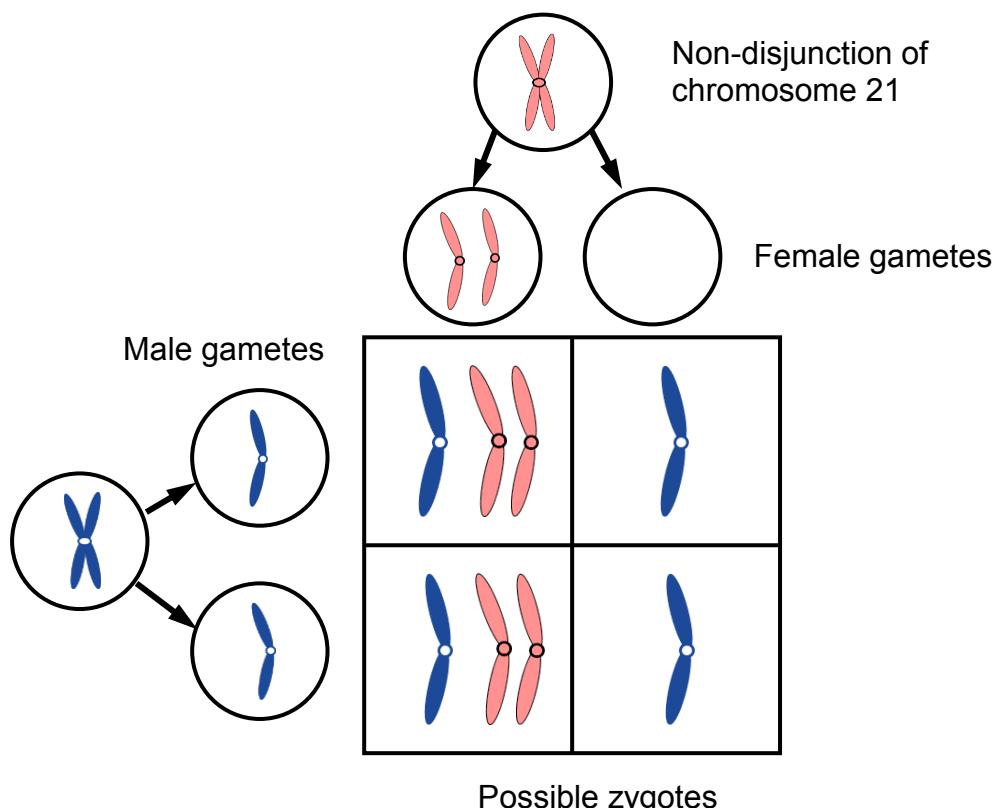


Figure 11: Punnet square showing chromosome aberrations

Biotechnology

For centuries, humans have used artificial selection to breed the best food crops, farm animals and pets (think of all the different varieties of dogs that exist today). With modern science, however, humans can manipulate the actual DNA of organisms.

Biotechnology is the use of organisms (e.g. bacteria) or biological processes to improve the quality of human life, as for example, in DNA profiling, genetic engineering, stem cell technology and cloning.

DNA profiling

DNA profiling was dealt with in Chapter 1. It is a form of biotechnology used for paternity testing, the identification of individuals, and for many other purposes.

Genetic engineering

Genetic engineering is used to alter the genome of a living cell for medical, industrial or agricultural purposes. This results in a **genetically modified organism** (GMO) or transgenic animal (animal with DNA from more than one species).

GMO's are used ...

- to breed more productive crops or animals so that more food can be made
- to produce drugs or hormones (e.g. insulin) which have fewer side-effects and is cheaper
- to 'infect' cells to cure diseases (gene therapy) such as brain tumours and cystic fibrosis

One process used to produce a GMO is **recombinant DNA technology**. It can be used to manufacture human insulin using *E. coli* bacteria. The process can be summarised as follows:

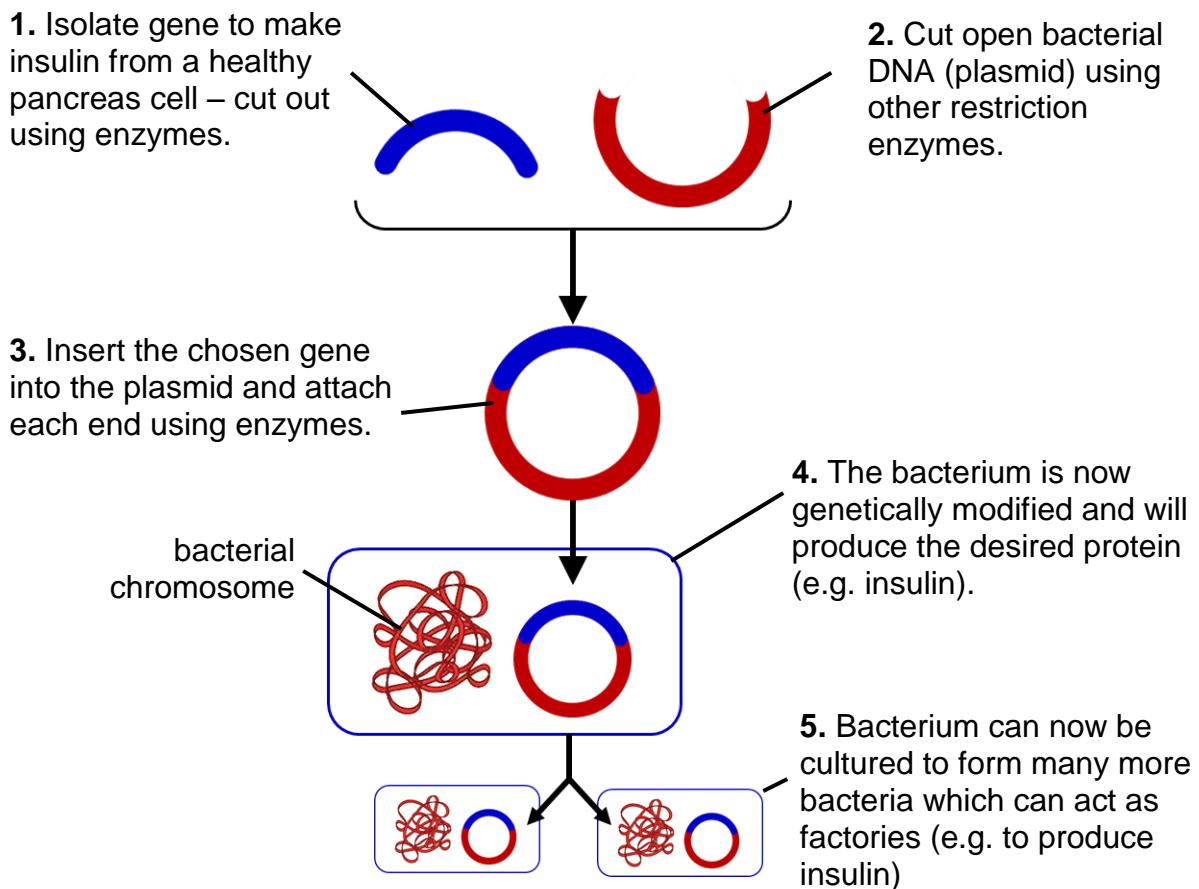


Figure 12: Recombinant DNA process

Advantages of genetically modified plants

- Pest resistance – the plants no longer taste good to insects.
- Herbicide tolerance – the crop plant is immune to poison, so large amounts can be used to kill the weeds.
- Disease resistance – these plants are hardy and do not get affected by diseases.
- Improved food quality – do not have damage due to pests or diseases so look good.
- Cold tolerance – rice and tobacco have been engineered to be unaffected by sudden drops in temperature.
- Drought or salinity tolerance – this helps plants grow in areas previously unsuitable for agriculture.
- Nutritionally enhanced – for example adding vitamin A to rice (a staple food in Asian countries) by introducing a gene from a daffodil.

- Incorporating vaccines into bananas/potatoes – this means that vaccines are made by the plant which can then be transported to countries easily without having to be refrigerated.

Disadvantages of GMO's

- GMO's contain glyphosate due to extensive spraying of herbicide.
- They are expensive so only 'rich' countries can benefit.
- The possibility of error is great as the process is complex.
- There has been no long-term safety testing as it is a relatively new technology.
- People may be allergic to the inserted gene e.g. brazil nut gene in soya beans.
- Widespread use of GMO crops may lead to a loss of biodiversity.
- New pathogens could be made for biological warfare.
- Ethically there is a fine line between what **can** be done and what **should** be done.

Only time will tell whether GMO's would solve the food security issues. Theoretically the potential benefits are huge, but there are significant risks. This also applies to the medical field and development of new drugs or ways to administer the drugs.

Stem cell technology

Stem cells are undifferentiated cells that have the ability to grow into any tissue in the body. They may be harvested from embryos left over after IVF treatment, from bone marrow and from blood in the umbilical cord. Skin and cartilage stem cells have also been used.

Notes about stem cells technology

- Embryonic stem cells are the most versatile as they have the ability to form any tissue. However, as the human embryos are killed, this is a controversial technology.
- Adult stem cells are much less controversial. Bone marrow has been used for a long time to treat cancers of the blood e.g. leukaemia, but other types of stem cell treatments are constantly being explored. Some of the procedures have involved:

- replacing dead cells in the heart after a heart attack
- growing skin tissue to treat burn victims
- growing nerve cells to treat spinal cord injuries and Parkinson's disease

However, a great deal more research is needed before these procedures are perfected. Parents who believe that there will be success in the future, are able to collect umbilical cord blood from their babies at birth. This blood can now be frozen and stored for future use. Although such facilities are available in South Africa, it is an expensive option.

Cloning

Cloning is the natural or artificial process of creating a genetically identical copy of an organism or biological material (e.g. tissue). The organism produced in this way is called a clone.

Cloning happens naturally when asexual reproduction takes place or a plant is self-pollinated or when identical twins are formed from a single zygote. These processes all give rise to individuals with DNA identical to that of the parent.

Biotechnology has enabled cloning to produce a new individual that is an exact copy of the organism from which the body cell was taken.

- In July 1996, Dolly the sheep, was the first cloned mammal using an adult cell, in this case a mammary gland cell.
- In April 2003, Futhi the cow, was the first cloned animal in South Africa and in this case a cell from the ear of a prize-winning dairy cow was used.



Figure 13: Dolly

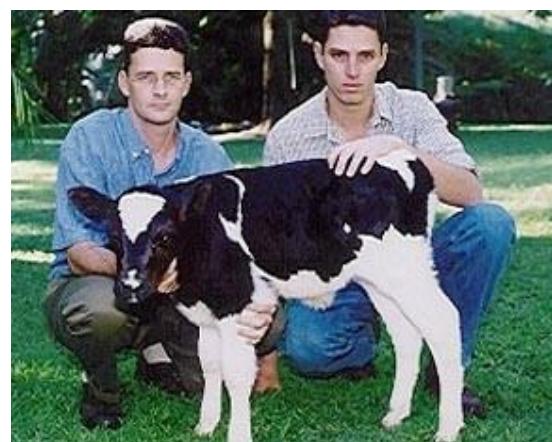


Figure 14: Futhi

- 2.1.1 How many of each of the following are represented in the diagram?
- Males (1)
 - Generations (1)
- 2.1.2 Give the
- phenotype of Jon (1)
 - genotype of Paul (1)
- 2.1.3 Both Lyall's parents can hear, yet he is deaf. Explain how deafness is inherited. (2)
- 2.1.4 Lyall marries a woman is homozygous dominant for hearing. Use a genetic cross to show the percentage chance of them having a deaf child. (7)
(13)
- 2.2 Tom and Maria have three children. One of the three children was adopted. A DNA profile for each member of the family was prepared to determine if Tom is the father of all three children (Anne, Mary and Steve). The DNA profiles are given below.
- | Tom | Maria | Anne | Mary | Steve |
|---|---|---|--|---|
|  |  |  |  |  |
- 2.2.1 Which one of the children was adopted? (2)
- 2.2.2 Explain your answer to question 2.2.1. (2)
(4)
- 2.3 Humans have different blood groups which are coded for by a number of alleles. Mary has genotype $I^A i$ and her son Joseph has blood type AB.
- How many alleles code for blood groups? (1)
 - Give all the possible genotypes for Joseph's father. (3)
(4)

- 2.4 Haemophilia is a genetic disorder caused by a recessive allele on the X-chromosome. A haemophiliac female marries a normal male. Explain why all their sons will be haemophiliacs. (4)
[25]

Question 3

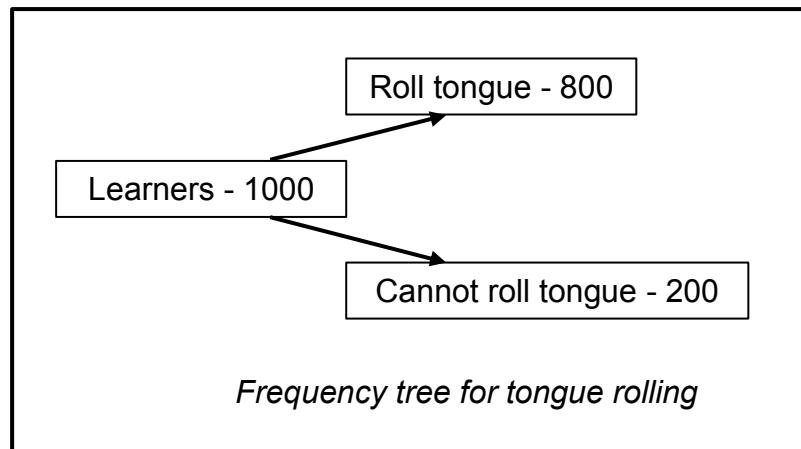
- 3.1 Coat colour in mice is controlled by two alleles, black (**B**) and grey (**b**). Tail length is controlled by two alleles, long (**T**) and short (**t**).

The Punnett square below shows a part of the cross between two mice. Genotype **(i)** has been left out.

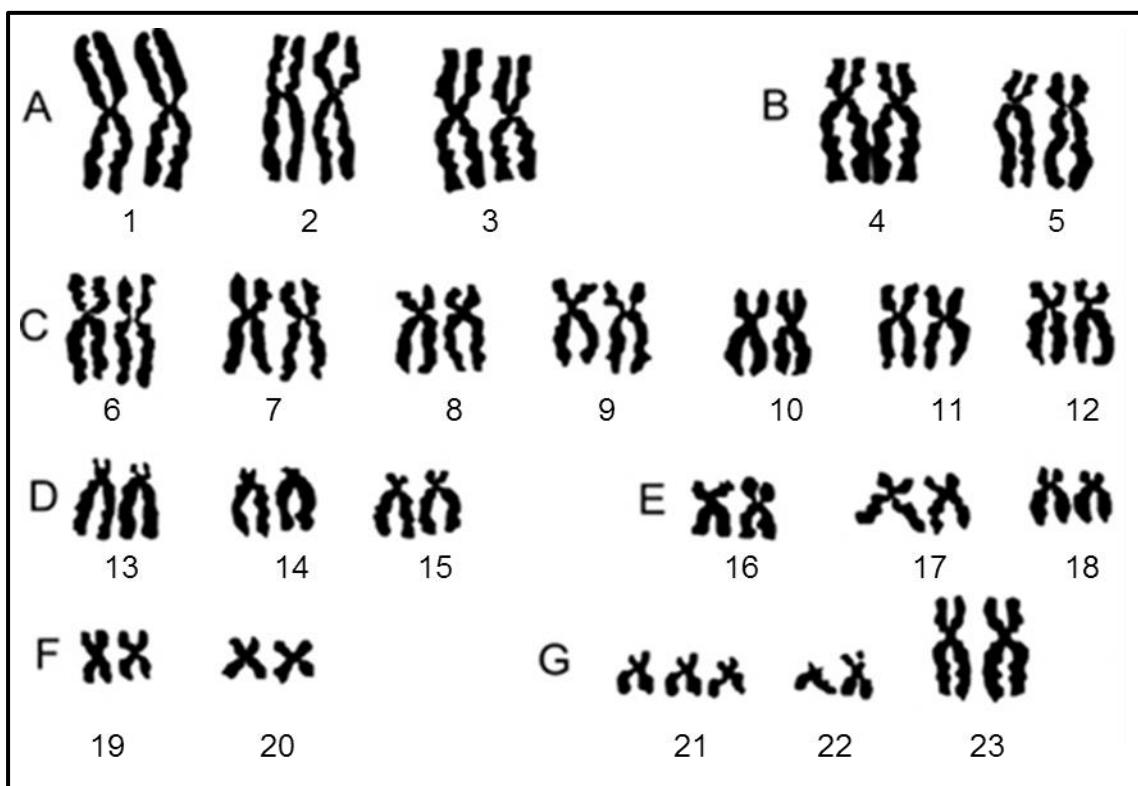
		Parent 1				
		Gametes	BT	Bt	bT	bt
Parent 2	Bt	BBTt	BBtt	BbTt	Bbtt	
	Bt	BBTt	BBtt	BbTt	Bbtt	
	Bt	BBTt	BBtt	(i)	Bbtt	
	Bt	BBTt	BBtt	BbTt	Bbtt	

- 3.1.1 Give the
- genotype of parent 1 (2)
 - phenotype of parent 2 (2)
 - genotype of offspring (i) (1)
- 3.1.2 What percentage of the offspring above is grey with short tails? (1)
- 3.1.3 State the genotypes of two gametes from the table above that will result in offspring that are heterozygous for both colour and length of tails if fertilisation occurs. (2)
(8)
- 3.2 A class of Grade 11 learners conducted an investigation to determine the frequency of dominant and recessive characteristics in their school. The characteristic investigated was the ability to roll one's tongue.

The results obtained were recorded in the frequency tree as shown below.



- 3.2.1 List three steps that the learners need to follow while planning this investigation. (3)
- 3.2.2 Use the data given in the frequency tree to plot a bar graph. (5)
- 3.2.3 Would you classify the ability to roll one's tongue as a continuous or discontinuous variation? (1)
- 3.2.4 Explain your answer to question 3.2.4 (2)
(11)
- 3.3 The diagram below is a representation of the chromosomes in a human cell.

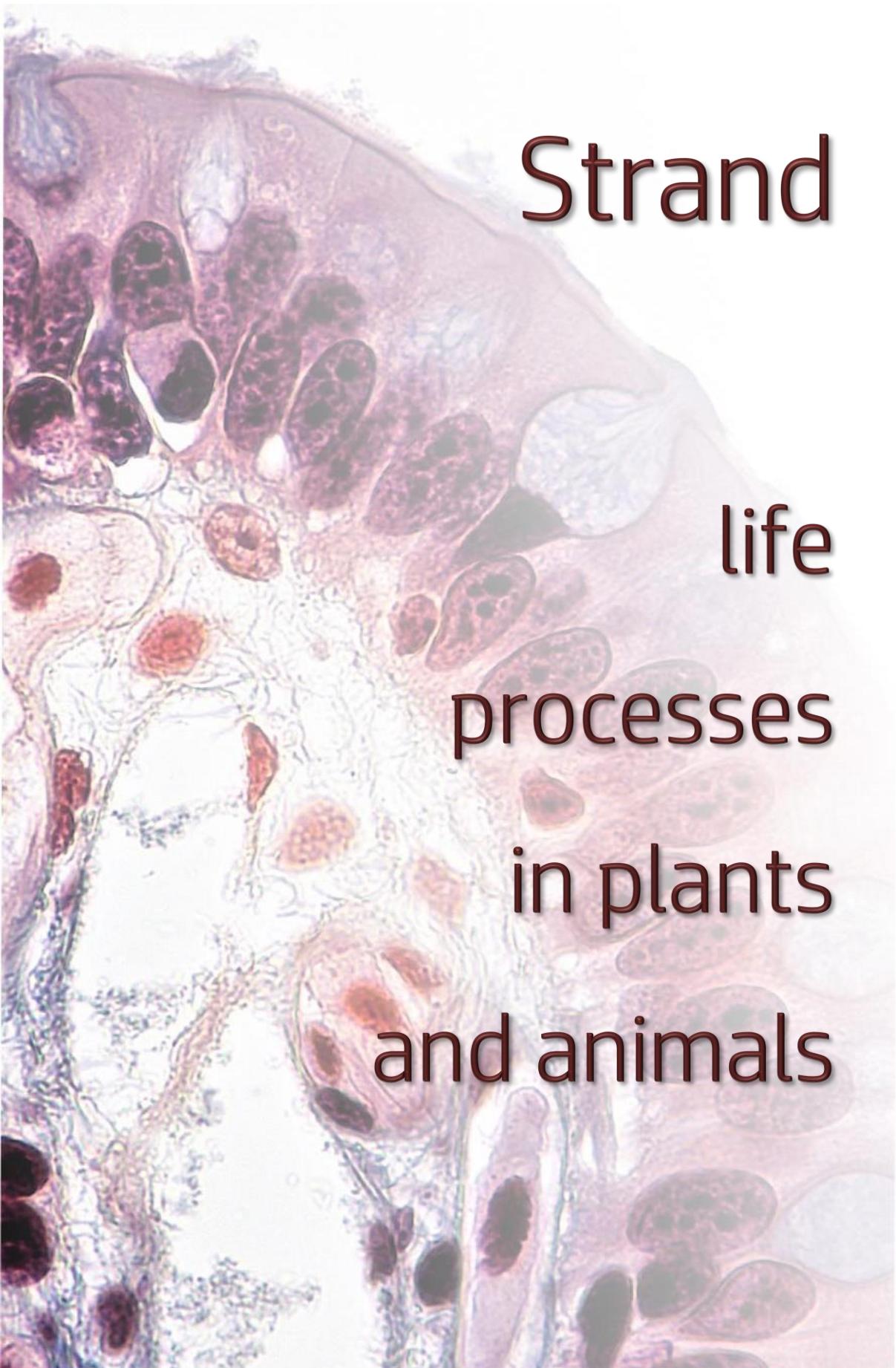


- 3.3.1 How many autosomes are in this cell? (1)
- 3.3.2 This individual is a female. Explain why this conclusion is made. (1)

- 3.3.3 What evidence is there to show that this individual has a genetic disorder? (1)
- 3.3.4 Identify the genetic order mentioned in question 3.3.3. (1)
- 3.3.5 Name the process that resulted in this genetic disorder. (1)
- (5)
[24]

Section B: [49]

Total Marks: [98]



Strand
life
processes
in plants
and animals

6: Human responses to the environment

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Human responses to the environment
The human nervous system
 Central nervous system & peripheral nervous system
 The central nervous system
 The brain
 The spinal cord
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 Synapse
 Activity 2: Neurons
Reflex action and reflex arc
 Activity 3: Reflex arc
 Activity 4: Practical investigation on reaction times
The peripheral nervous system
 Somatic nervous system
 Autonomic nervous system
Disorders, injuries and effects of drugs on the nervous system
 Alzheimer's disease
 Multiple sclerosis

Injuries
Effects of drugs
Sense organs
 The human eye
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CHAPTER 6: HUMAN RESPONSES TO THE ENVIRONMENT

Introduction

Humans respond to the environment in various ways to maintain homeostasis, function efficiently and protect themselves from danger. The human nervous system with its receptor organs allows the body to respond.

Key terminology

receptor	structure that receives a stimulus and converts it into an impulse
effector	gland or organ that brings about a response to stimuli received by the body
stimulus	detectable change in the internal or external environment
impulse	electrical signal created by receptor organs in response to stimuli
transmit	to send something from one place to another
autonomic nervous system	controls our involuntary bodily functions; it is divided into the parasympathetic and sympathetic nervous system
peripheral nervous system	consists of nerves that extend outside the central nervous system (brain and spinal cord)

Human responses to the environment

In order to survive all organisms must be able to respond to the environment. To achieve this, humans have two co-ordinating systems which allow them to adapt to changes within the environment and maintain their internal conditions. A co-ordinating system consists of receptors and effectors which respond to stimuli and allow for changes to take place. These two systems are the nervous system and the endocrine system. The nervous system is made up of nerves and is a fast responding system. The endocrine system involves hormones which are released into the blood and is a much slower responding system.

The human nervous system

Key terminology

cranium	part of the skull that contains and protects the brain
meninges	protective membranes surrounding the brain & spinal cord
cerebrospinal fluid	fluid around the brain and spinal cord to aid in protection
grey matter	part of the brain and spinal cord consisting of cell bodies and dendrites
white matter	part of the brain and spinal cord consisting of myelinated axons
neuron	specialised nerve cells which transmits nerve impulses
dendrites	fibres that transmit impulses to a cell body in a neuron
nerve	bundle of neurons
synapse	the gap between the axon of one neuron and the dendrite of another
neurotransmitter	chemicals which carry impulses across the synapse
homeostasis	the tendency of living organisms to maintain their internal environment constant within narrow limits irrespective of changes in the external environments
Alzheimer's disease	disease caused by nerve defects usually in older people and characterised by memory loss and confusion
multiple sclerosis	disease cause by damage to the myelin sheath of neurons and characterised by physical and mental disabilities

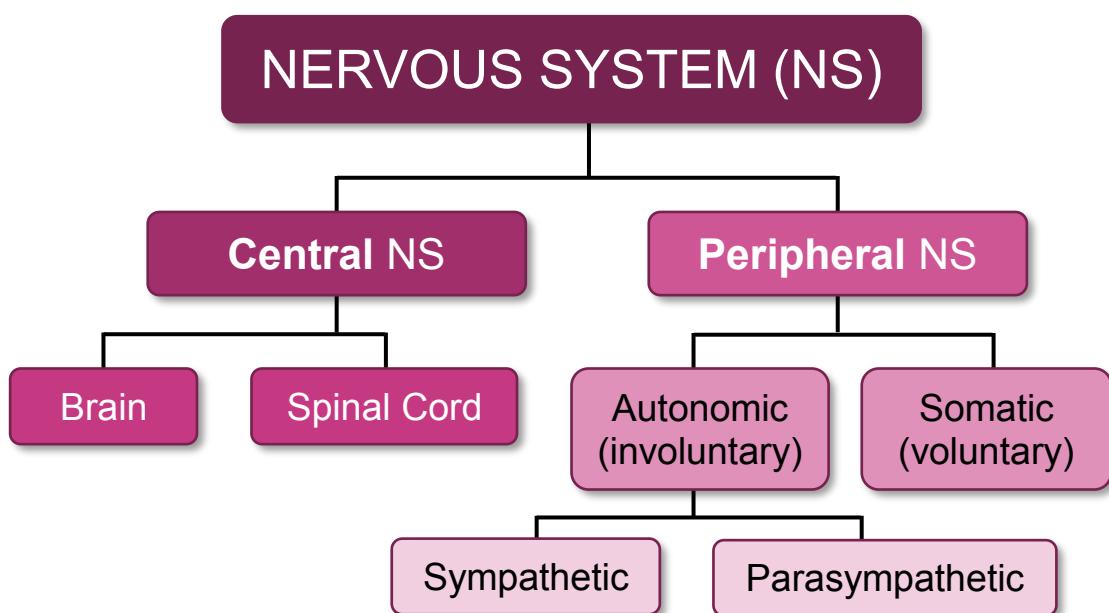


Figure 1: The human nervous system

Central nervous system & peripheral nervous system

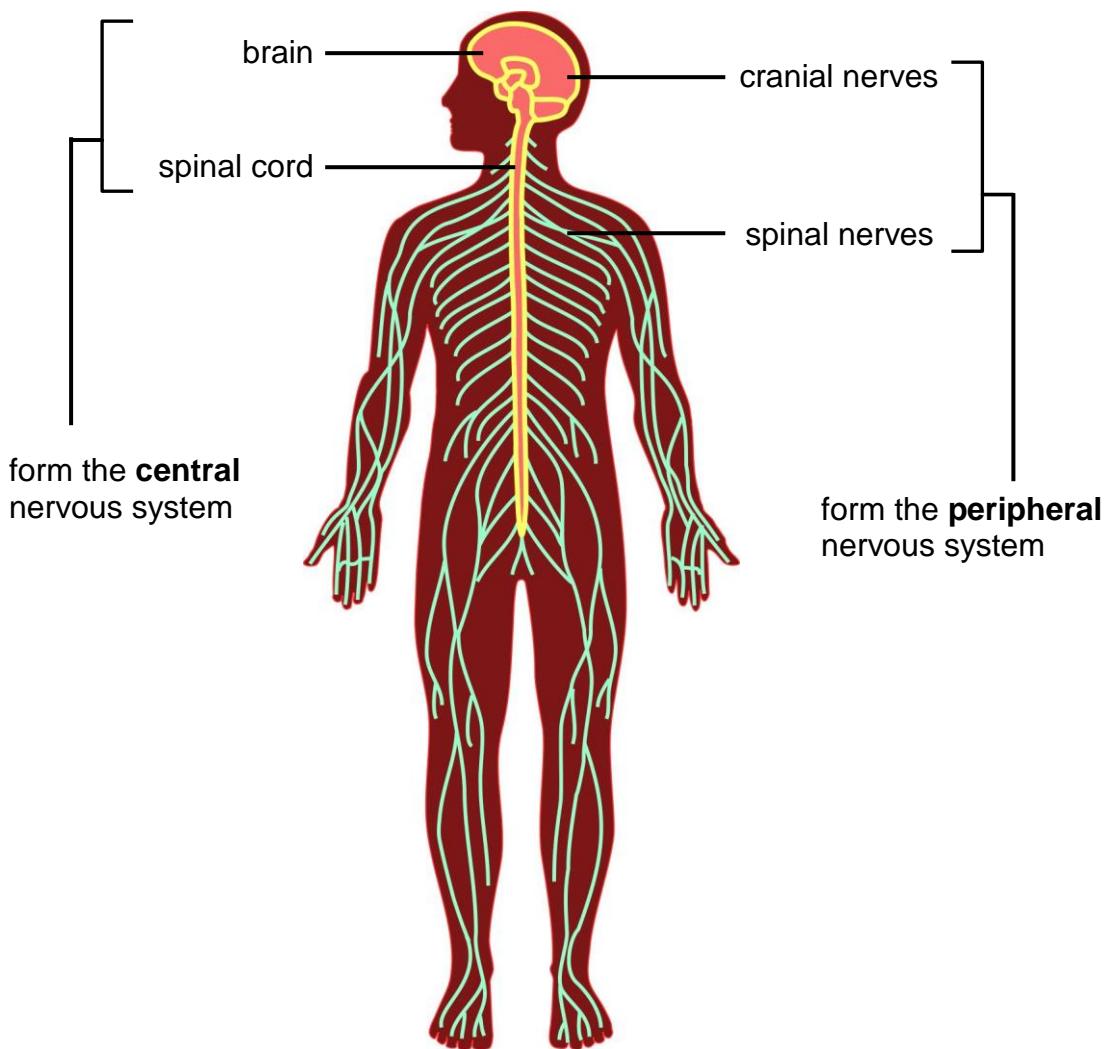


Figure 2: The central nervous system and peripheral nervous system

As illustrated in Figures 1 and 2 above, the human nervous system may be divided into two parts.

- The central nervous system which consists of the brain and spinal cord.
- The peripheral nervous system which consists of an autonomic part which is involuntary (one that can't be controlled) and a somatic part which is voluntary (under conscious control).

Examples of a voluntary action would be running, jumping, eating or walking. Involuntary actions, or reflex actions, include breathing and sneezing.

- The autonomic part may be further divided into the sympathetic and the parasympathetic systems, which work antagonistically (in an opposing manner).

Humans experience many changes throughout the day to which they must respond. These changes are known as stimuli. A stimulus may be internal (e.g. body temperature, water, sugar levels, carbon dioxide and oxygen concentrations) or it may be external (e.g. pain, environmental temperature, danger). Regardless of whether a stimulus is internal or external, we must be able to respond.

The central nervous system

The central nervous system is made up of the brain and spinal cord.

The brain

The brain is made up of delicate nervous tissue which cannot repair itself. The brain is of vital importance because it controls all functioning of the human body. For this reason, it is well protected in three ways:

- It is inside a bony **cranium**
- It is surrounded by three membranes called the **meninges**
- It is cushioned by a fluid called **cerebrospinal fluid**

Figure 3 shows various important structures of the brain, and their location relative to each other.

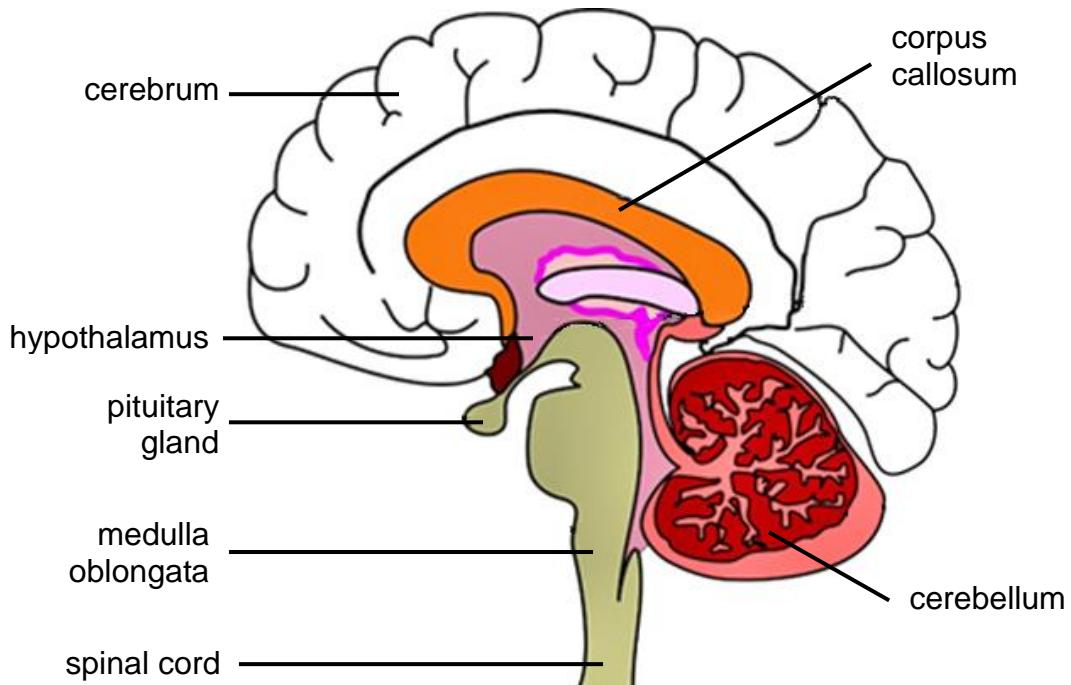


Figure 3: The structure of the brain

Table 1 below shows the location and function of various parts of the brain.

Table 1: The structure and function of various parts of the brain

Structural adaptation	Function
cerebrum	
<ul style="list-style-type: none"> the largest part of the brain divided into two hemispheres (left and right) which are connected by the corpus callosum 	<ul style="list-style-type: none"> controls voluntary functions (walking, speaking, writing) receives and interprets sensations from sense organs (hearing, sight, feeling, taste, smell) higher thought processes (memory, intelligence, reasoning) to allow communication between the two halves of the brain
cerebellum	
<ul style="list-style-type: none"> second largest part of the brain located behind and below the cerebrum 	<ul style="list-style-type: none"> co-ordinates skeletal muscles to bring about balance while moving, as in walking or running maintains balance and posture maintains muscle tone
medulla oblongata	
<ul style="list-style-type: none"> lower part of the brain continues down into the body as the spinal cord 	<ul style="list-style-type: none"> controls breathing, peristalsis, heartbeat, swallowing transmits impulses from the spinal cord to the brain controls less important reflexes: blinking, coughing, sneezing, vasodilation, vasoconstriction and salivating
hypothalamus	
<ul style="list-style-type: none"> a small section of the brain just above the pituitary gland 	<ul style="list-style-type: none"> a control centre for things such as hunger, thirst, sleep, body temperature, emotions

Parts of the brain: <https://www.youtube.com/watch?v=-nH4MRvO-10&t=86s>

The spinal cord

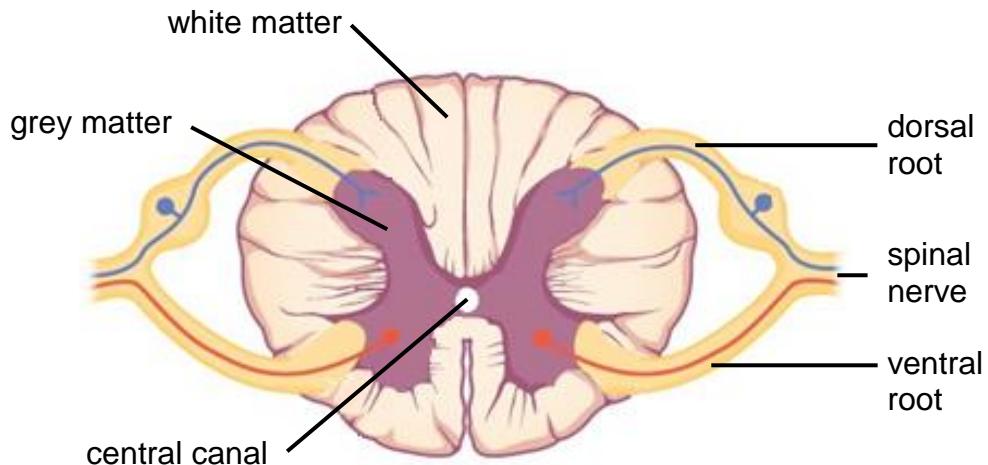


Figure 4: Internal structure of the spinal cord.

The spinal cord is made up of delicate nervous tissue which cannot repair itself. It is therefore protected by:

- 33 vertebrae (bone) with discs of cartilage between them to act as shock absorbers
- three membranes called the meninges
- cerebrospinal fluid

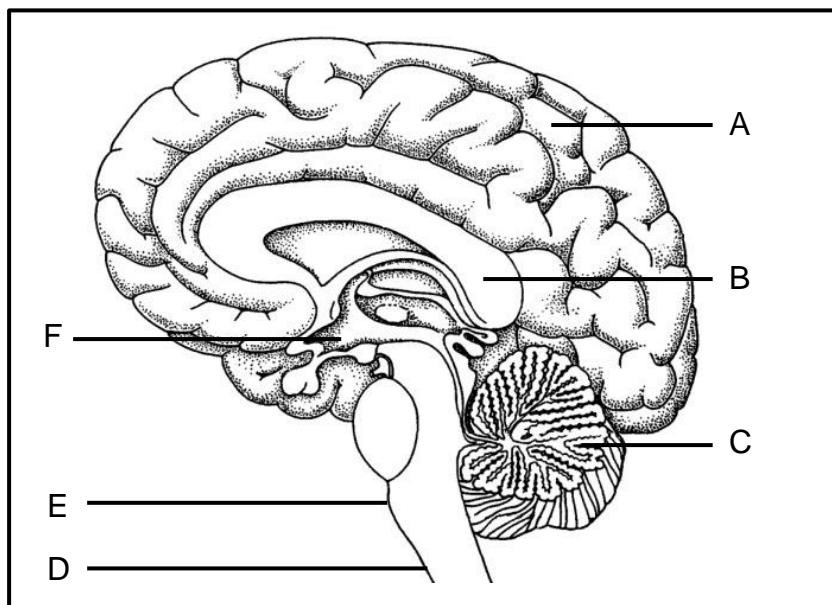
The spinal cord passes from the medulla oblongata to the lumbar region in the lower back. Thirty-one pairs of spinal nerves are attached to the spinal cord through the openings between the vertebrae. Each spinal nerve has a dorsal root which enters and a ventral root which leaves the spinal cord.

The spinal cord has the following functions:

- transmits impulses from receptors to the brain and from the brain to the effectors
- contains reflex centres that function automatically to protect the body

Activity 1: The central nervous system

Study the diagram below representing the human brain.



1. Give the names of the parts labelled A to C. (3)
 2. Give the letter and the name of the part responsible for:
 - a) co-ordinating all voluntary movements (2)
 - b) memorising a cell phone number (2)
 3. Explain two functions of parts:
 - a) E (2)
 - b) F (2)
 4. Provide two ways in which part D is protected. (2)
 5. Which functions of the body might be affected if the lower back part of the head receives a significant impact in a car accident? (2)
- (15)

Neurons

Neurons are the structural units of the nervous system. Neurons are specialised cells which connect the brain and spinal cord to all other parts of the body.

As illustrated in Figure 5 below, neurons have the following structures:

- dendrites – transmitting impulses towards the cell body
- a cell body (with nucleus) – controlling the metabolism of the cell
- an axon – transmitting impulses away from the cell body

- a myelin sheath – insulating the axon and speeding up transmission of impulses; the myelin sheath is enclosed by a membrane called the neurilemma which helps to repair damaged neurons

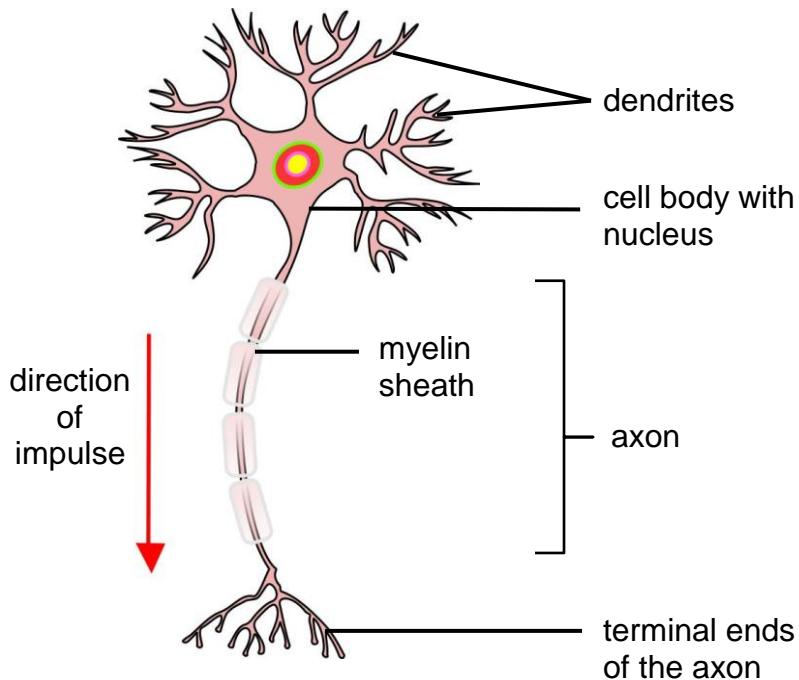


Figure 5: Structure of a motor neuron

There are three types of neurons: sensory neurons, interneurons and motor neurons. These are illustrated in Figures 6A to 6C.

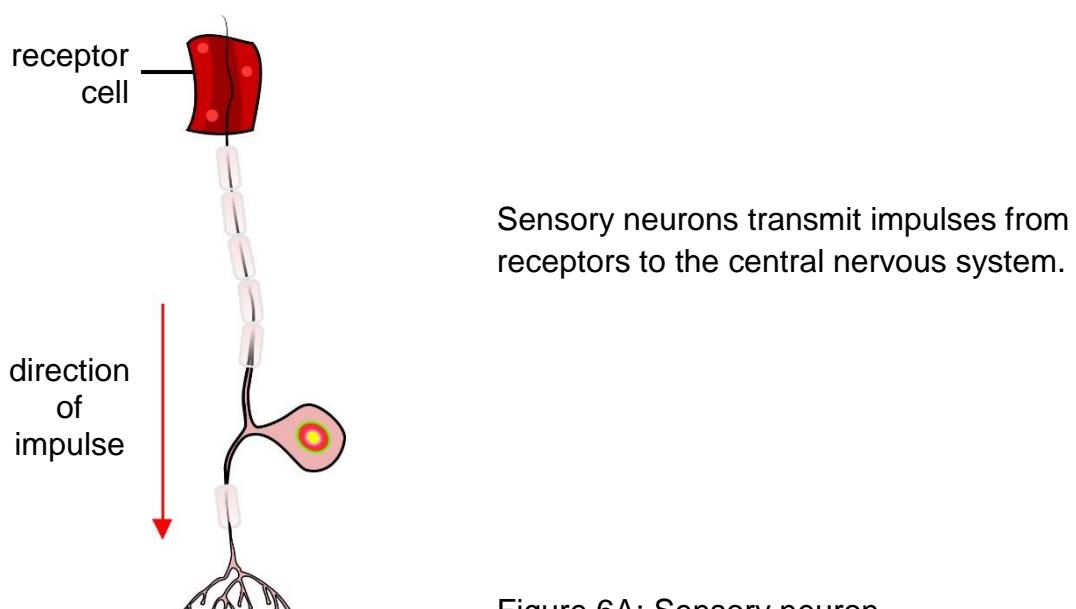
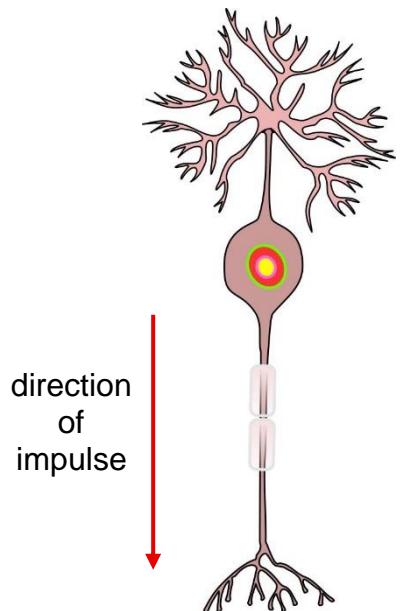
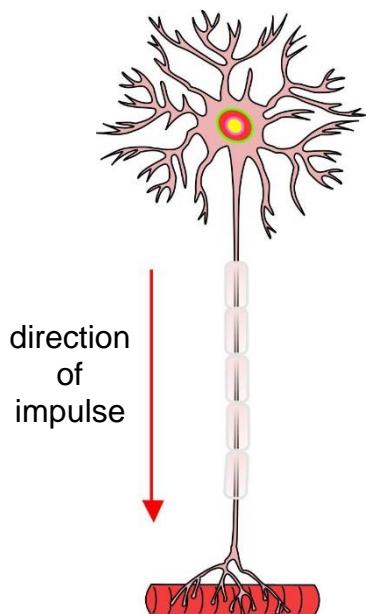


Figure 6A: Sensory neuron



An interneuron connects a sensory neuron to a motor neuron in the central nervous system.

Figure 6B: Interneuron



Motor neurons transmit impulses from the central nervous system to the effectors (muscles and glands) in the body.

Figure 6C: Motor neuron

Synapse

A synapse (see Figure 7 below) is a functional connection between the axon of one neuron and the dendrites of another neuron. The connection is established by neurotransmitters, chemicals that move across the gap between axon and dendrite to transmit the impulse.

Significance of a synapse

- It ensures that the impulse moves in one direction only
- It prevents continuous stimulation of the neurons

- It ensures that the impulse is transmitted from the sensory neuron to the motor neuron

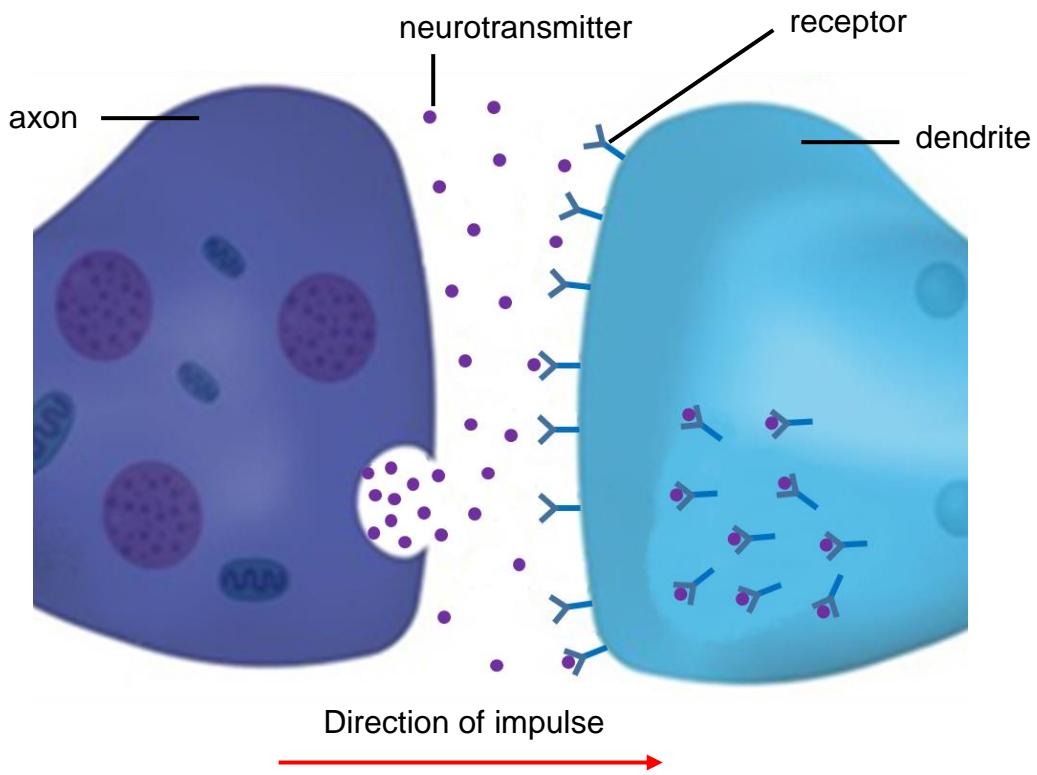


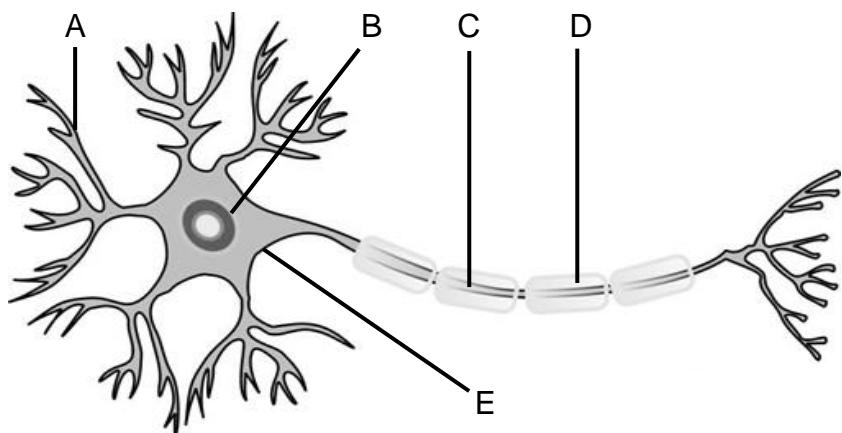
Figure 7: A synapse

Types of neurons:

<https://www.youtube.com/watch?v=n0Zc01e1Frw&list=PLW0gavSzhMIQYSpKryVcEr3ERUp5SxHI0&index=48>

Activity 2: Neuron structure

The diagram below represents the structure of a neuron.



1. Name the type of neuron shown in the diagram above. (1)
 2. Identify the parts A to E. (5)
 3. Provide the functions of the following parts:
 - a) C (1)
 - b) D (1)
 4. Describe the pathway of an impulse through the neuron shown above. (1)
 5. Draw a fully labelled diagram to show the structure of a neuron which receives impulses and transmits them to the central nervous system. (4)
- (13)

Reflex action and reflex arc

- A **reflex action** is a quick, automatic response to a stimulus. Examples: knee-jerk, sneezing and quickly removing a body part away from danger to respond to pain.
- A **reflex arc** is the pathway along which an impulse is transmitted to bring about a response to a stimulus during a reflex action.

Significance of Reflex action

- The reflex action allows for a quick response, without thinking about it, to prevent damage to the body.

The diagram below (Figure 8) can be used to explain how a reflex action takes place. The arrows in the diagram show the reflex arc.

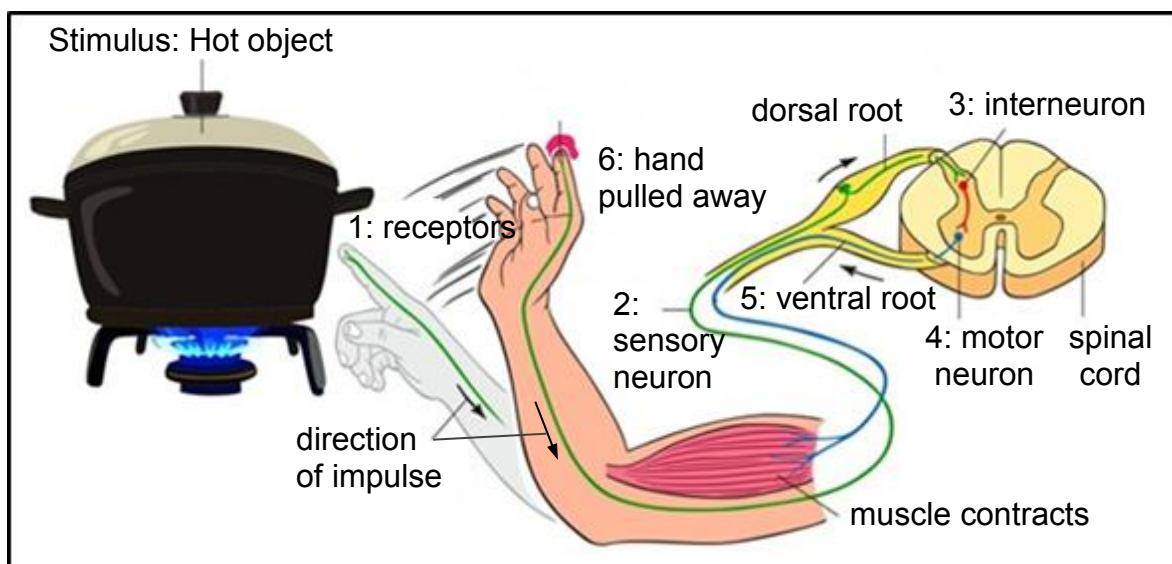


Figure 8: A reflex action

The reflex action of a person touching a hot pot:

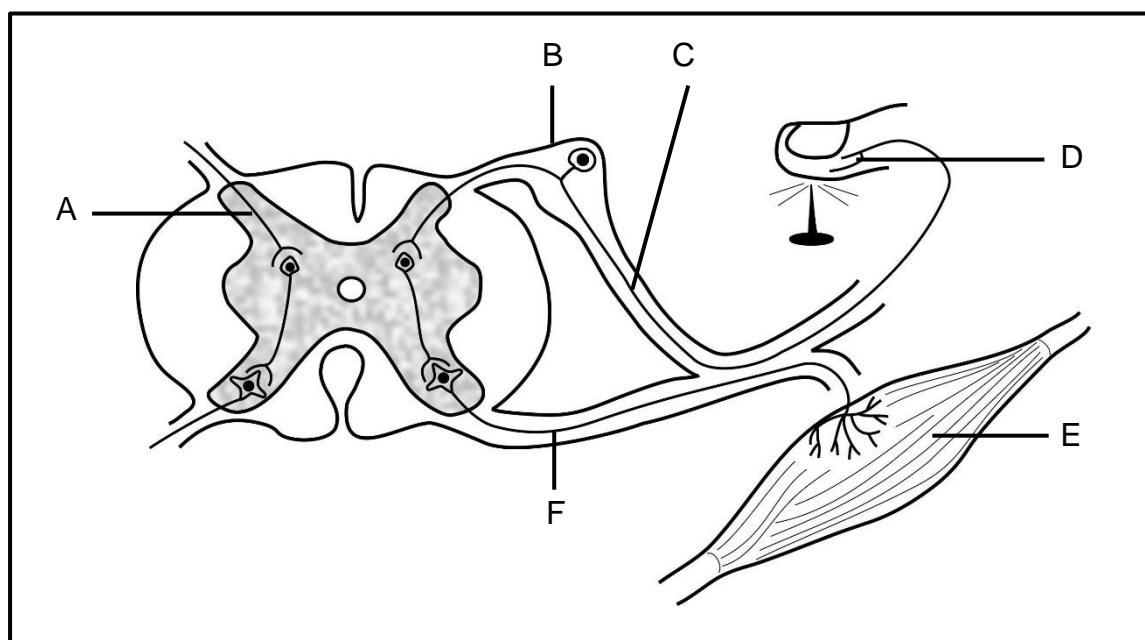
1. The stimulus is detected by receptors and converted into a nerve impulse.
2. The nerve impulse is transmitted along the sensory neuron through the dorsal root to the spinal cord.
3. The impulse is transmitted from the sensory neuron to an interneuron in the spinal cord.
4. The impulse is transmitted from the interneuron to a motor neuron in the spinal cord.
5. The impulse exits the spinal cord through the ventral root and is transmitted along the axon of the motor neuron to the effector organs (muscle; this causes the muscles in the arm to contract).
6. The hand pulls away from the stimulus quickly.

This reflex action does not include the brain. Impulses will reach the brain after the reflex arc is complete and pain will be felt.

How a reflex arc functions: <https://www.youtube.com/watch?v=Nn2RHLWST-k>

Activity 3: Reflex arc

The diagram below shows the reflex arc. Study the diagram, then answer the questions below.



- Give only the letter of the part that represents the:
 a) effector (1)
 b) interneuron (1)
 c) sensory neuron (1)
 - State one function of each of the following parts:
 a) D (1)
 b) F (1)
 - Explain why the brain is not initially involved in the reflex action. (3)
 - Explain the effect on the body if the following parts were cut:
 a) C (4)
 b) F (4)
- (16)

Activity 4: Practical investigation on reaction times

Thando conducted an experiment to determine which gender has the faster reaction time amongst his classmates. Out of 15 learners in his class he randomly selected a sample of 5 girls and 5 boys. The following steps were followed for each member of the sample during the experiment:

- Thando held a meter ruler between his thumb and index finger just above the 100 cm mark.
- The pupil placed the thumb and index finger on either side of the meter ruler at the 0 cm mark, with only the thumb touching it.
- As Thando dropped the meter ruler the pupil caught it by closing the thumb and forefinger.
- During each trial Thando recorded the distance at which the meter ruler was caught.
- The procedure was repeated five times for each pupil.

The table below shows the average distance at which the meter ruler was caught by 5 boys and 5 girls over five trials.

Average distance at which the meter ruler was caught over 5 trials (cm)

Boys	average distance (cm)	Girls	average distance (cm)
Boy 1	5,8	Girl 1	4,8
Boy 2	5,0	Girl 2	4,7
Boy 3	4,9	Girl 3	4,2
Boy 4	4,8	Girl 4	4,0
Boy 5	4,6	Girl 5	3,9
overall average	5,02	overall average	4,32

1. Identify the:
 - a) independent variable of the experiment, and (1)
 - b) dependent variable of the experiment. (1)
2. Give two reasons why the results of this experiment may be regarded as reliable. (2)
3. Mention two factors which should be kept constant during the experiment. (2)
4. Thando's initial hypothesis was rejected on the basis of the results obtained. Suggest what Thando's initial hypothesis could have been. (2)
(8)

The peripheral nervous system

The peripheral nervous system consists of all the nerves found outside of the central nervous system. The peripheral nervous system consists of 12 pairs of cranial nerves which are connected to the brain and 31 pairs of spinal nerves which are connected to the spinal cord.

The peripheral nervous system can be divided into two parts:

- the somatic nervous system which controls voluntary muscles
- the autonomic nervous system which controls involuntary muscles

The peripheral nervous system has the following functions:

- transmits impulses from the receptors to the central nervous system via the sensory neurons
- transmits impulses from the central nervous system to the effectors via the motor neurons

Somatic Nervous System

The somatic nervous system controls voluntary (skeletal) muscles. The nerves in this system allow the body to react to changes in the external environment.

Autonomic Nervous System

The autonomic nervous system controls involuntary actions. The nerves in this system allow the body to react to changes in the internal environment so that homeostasis can be maintained.

The autonomic nervous system can be subdivided into the sympathetic nervous system and the parasympathetic nervous system. Each organ in the human body is supplied by two both types of nerves: sympathetic nerves and parasympathetic nerves.

The sympathetic nervous system is responsible for the fight or flight function in emergency situations while the parasympathetic system restores the body to a normal state after an emergency situation. The two systems thus work antagonistically to each other.

Table 2 below shows how these systems work in opposition to bring about homeostasis.

Table 2: Effects of the sympathetic and parasympathetic nervous systems

Sympathetic system	Parasympathetic system
increases heart rate	decreases heart rate
constricts blood vessels in the skin (vasoconstriction)	dilates blood vessels in skin (vasodilation)
increases blood pressure	decreases blood pressure
widens bronchioles	narrows bronchioles
decreases peristalsis	increases peristalsis
causes relaxation of the bladder wall	causes contraction of the bladder wall
stimulates sweat secretion	no effect
dilates pupils	constricts pupils
stimulates secretion of adrenalin	no effect

Note: Adrenalin is a very important hormone that will be discussed in a later chapter. It works with the sympathetic nervous system to prepare the body for dangerous or stressful situations.

Disorders, injuries and Effects of Drugs on the Nervous system

There are many disorders and injuries which can affect the nervous system. The use of drugs can temporarily or permanently affect the functioning of the nervous system. How much the nervous system will be effected depends on many things.

Alzheimer's disease

Alzheimer's disease is a neurodegenerative disease which means there is progressive brain cell death that occurs over time. The disease is irreversible.

It is not yet known what causes the disease. In most cases, the first symptoms appear after the age of 60 but it has been seen in people as young as 40.

There is no known cure but the symptoms can be managed.

Symptoms:

- memory loss
- confusion

Multiple sclerosis

Multiple sclerosis is a disease that affects young adults between the ages of 20 and 40. The body's immune system attacks the myelin sheath covering neurons which prevents them from functioning properly. The reason why this happens is unknown. There is no known cure for this disease. The symptoms can be managed with medication.

Symptoms:

- loss of speech and vision
- difficulty walking
- pain
- fatigue
- memory loss

Injuries

There are many causes of brain and spinal cord injuries, but the most common cause is motor vehicle accidents.

Brain injuries can affect movement, memory and speech but this depends on the part of the brain that is injured. In severe cases, they may lead to long-term mental health issues.

A damaged spinal cord could result in paralysis and the inability to move or feel anything below the point of injury.

There is no way of regenerating damaged nerve tissue, however regular occupational and physical therapy may allow patients to regain some feeling and function.

Medical researchers claim that stem cell therapy may in future be used to repair spinal cord injuries, degenerative diseases such as multiple sclerosis and Parkinson's disease.

Effects of drugs (Not examinable)

Drugs change the way in which impulses are transmitted in the central nervous system, mainly by affecting how quickly or slowly the neurotransmitters can pass across the gap in the synapse. Depending on the type of drug used (e.g.: marijuana, ecstasy, heroine, tik) either the sympathetic or the parasympathetic nervous system will become more stimulated. This can lead to side-effects such as:

- reduced co-ordination
- increased heart rate
- increased blood pressure
- decreased appetite
- hallucinations and paranoia

Sense Organs

Sense organs contain a high concentration of receptor cells which are able to detect stimuli to bring about a response. We will study the eye and the ear.

The human eye

Key terminology

rods	receptor cells found in the retina of the eye which are sensitive to dim light and help to distinguish between black and white
cones	receptor cells found in the retina of the eye which are sensitive to bright light and help to distinguish between different colours
pupil	central opening within the iris which allows light to enter

pupillary mechanism	regulation of the pupil size to control the amount of light entering the eye
accommodation	the ability of the lens of the eye to alter its shape for clear vision when viewing both near and distant objects
field of vision	the area that one eye can see
convex	a shape which curves outwards, thicker in the middle than the edges
concave	a shape which curves inwards, thinner in the middle than the edges

The receptors that detect light are called rods and cones. These are situated in the retina at the back of the human eye. The eyes are found as a pair positioned at the front of the skull. The field of vision from each eye overlaps which allows a slightly different image of the same object from each eye. Two types of imaging occur as a result:

- **Binocular vision:** vision using two eyes with overlapping fields of view so that the separate images are combined and interpreted as one image by the brain.
- **Stereoscopic vision:** the ability to form three dimensional images which provides the ability to judge distance, depth and the size of an object.

Structure of the eye

Figure 9 below gives a view of an eye from the front.

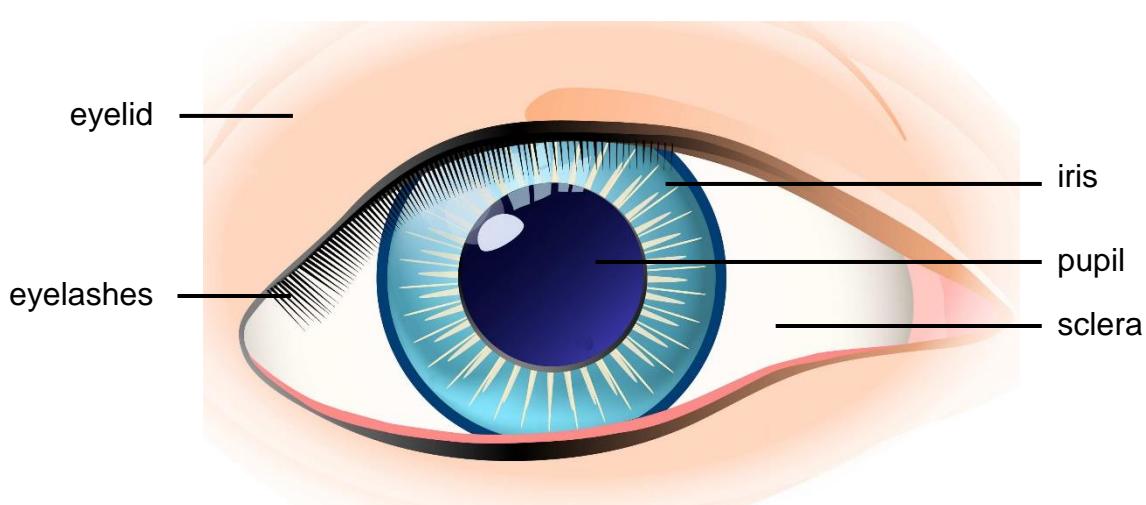


Figure 9: Front view of human eye

The eyeballs lie inside a bony cavity. Connective tissue and fat surround the eyeball to protect it from mechanical injury. The front of the eye is protected by eyelids, which have eyelashes to stop foreign particles from entering the eye. The coloured part of the eye is known as the iris. The opening in the centre of the iris is the pupil. The sclera is a white outer covering layer.

The internal eye can be divided into three layers: the sclera, choroid and the retina. These layers, and other parts of the internal eye are shown in Figure 10 below.

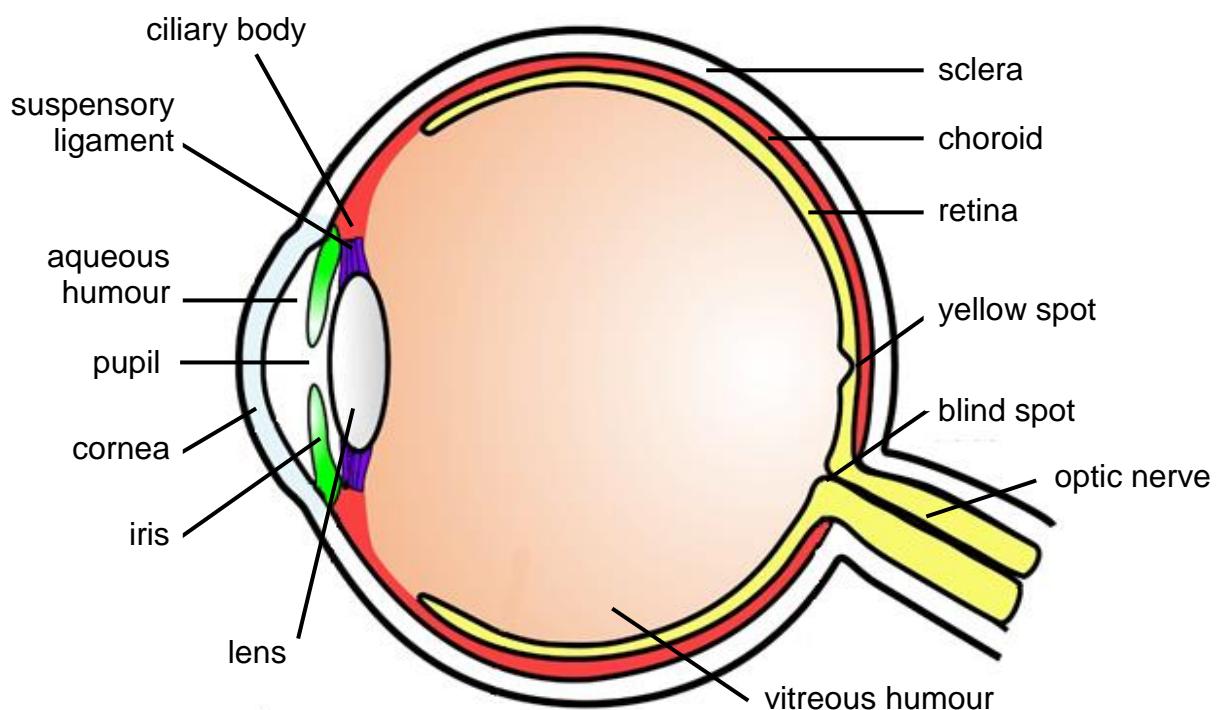


Figure 10: A longitudinal section of the eye

The structural adaptation and function of the parts of the eye shown above are laid out in Table 3 below.

Table 3: Structural adaptations and functions of various parts of the eye

Structural adaptations	Functions
sclera	
<ul style="list-style-type: none"> • tough, white inelastic layer that covers eye towards the posterior • adaptation: protection of the eye because it is inelastic 	<ul style="list-style-type: none"> • protects the inner structures of the eye • maintains the shape of the eye

cornea	<ul style="list-style-type: none"> transparent continuation of the sclera in the front of the eye which is convex (bulgy) adaptation: transparency allows light to pass through; the convex shape causes refraction (bending) of the incoming light 	<ul style="list-style-type: none"> allows light to pass through causes refraction (bending) of the incoming light to create an image on the retina
choroid	<ul style="list-style-type: none"> a dark coloured layer which contains blood vessels and pigments 	<ul style="list-style-type: none"> pigments absorb light to prevent the reflection of light blood vessels supply nutrients and oxygen to the cells of the retina
ciliary body	<ul style="list-style-type: none"> contains ciliary muscles and it is the thickened part at the anterior of the choroid 	<ul style="list-style-type: none"> ciliary muscles contract or relax to alter the tension on the suspensory ligaments
suspensory ligaments	<ul style="list-style-type: none"> ligaments attached to the ciliary body 	<ul style="list-style-type: none"> suspensory ligaments hold the lens in position during accommodation tension on the suspensory ligaments changes to alter the shape of the lens
iris	<ul style="list-style-type: none"> the coloured portion of the eye, with an opening at its centre, called the pupil; the iris contains two types of muscles to control the size of the pupil 	<ul style="list-style-type: none"> controls the amount of light entering the eye through the pupillary mechanism
retina	<ul style="list-style-type: none"> inner layer of the eye which contains the rods and cones that are sensitive to light 	<ul style="list-style-type: none"> rods respond to low intensity light, provide night vision as well as peripheral vision cones respond to bright light and provide sharp, clear colour vision neurons carry impulses from the rods and cones through the optic nerve to the cerebrum
yellow spot	<ul style="list-style-type: none"> small indentation at the back of the eyeball containing the most cones 	<ul style="list-style-type: none"> area of clearest vision
blind spot		

<ul style="list-style-type: none"> small area on the retina, below the yellow spot; contains no rods or cones, and hence no vision area where the blood vessels enter the eye area where the optic nerve leaves the eye 	<ul style="list-style-type: none"> area of no vision inner parts of the eye are supplied with oxygen and nutrients impulses are transmitted to the cerebrum for interpretation
lens	
<ul style="list-style-type: none"> elastic and biconvex structure behind the pupil of the iris; held in place by suspensory ligaments transparent 	<ul style="list-style-type: none"> changes shape to allow the eye to focus on near and distant objects allows light to pass through
aqueous humour	
<ul style="list-style-type: none"> watery fluid found in the space between the cornea and the lens 	<ul style="list-style-type: none"> maintains the shape of the cornea plays a small role in the refraction of the incoming light
vitreous humour	
<ul style="list-style-type: none"> jelly-like substance found behind the lens 	<ul style="list-style-type: none"> maintains the shape of the eyeball plays a small role in the refraction of the incoming light

Structures of the human eye: <https://www.youtube.com/watch?v=syaQgmxb5i0>

Accommodation

Accommodation is the ability of the eye to alter the shape of the lens to ensure that a clear image always falls on the retina whether the object is near or distant. The process is explained in Table 4 below, and illustrated in Figures 11A for near vision and 11B for distant vision.

Table 4: Accommodation of the human eye

Near vision (less than 6 m from the object)	Distant vision (more than 6 m from the object)
<ul style="list-style-type: none"> ciliary muscles contract suspensory ligaments slacken (loosen) tension on the lens decreases lens becomes more convex (bulgy) this causes light rays to bend more a clear image is focused on the retina 	<ul style="list-style-type: none"> ciliary muscles relax suspensory ligaments tighten (become taut) tension on the lens increases lens becomes less convex (flatter) this causes light rays to bend less a clear image is focused on the retina

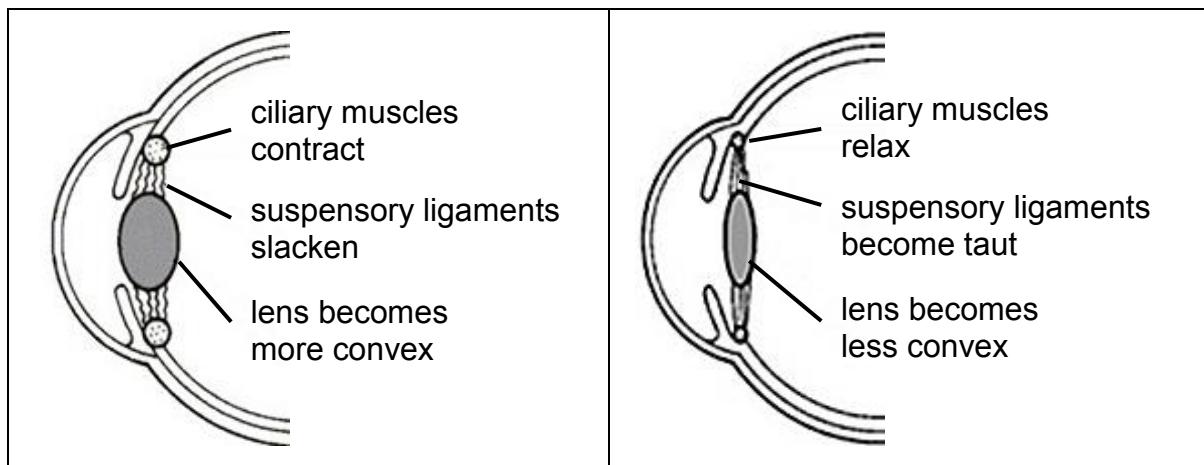


Figure 11A: Near vision

Figure 11B: Distant vision

Pupillary mechanism

Pupillary mechanism refers to the process by which the diameter of the pupil is altered to control the amount of light entering the eye. The intensity of the light is the stimulus that changes the size of the pupil. The iris controls the amount of light entering the pupil. It has circular and radial muscles which act antagonistically to change the size of the pupil.

Table 5 and Figures 12A and 12B below show the pupillary mechanism as it occurs in bright light and in dim light conditions.

Table 5: Pupillary mechanism

Bright light conditions	Dim light conditions
<ul style="list-style-type: none"> radial muscles cause iris to relax circular muscles of the iris contract the pupil constricts (gets smaller) the amount of light entering the eye is reduced 	<ul style="list-style-type: none"> radial muscles cause iris to contract circular muscles of the iris relax the pupil widens (gets bigger) the amount of light entering the eye is increased

Figure 12A: Bright light conditions

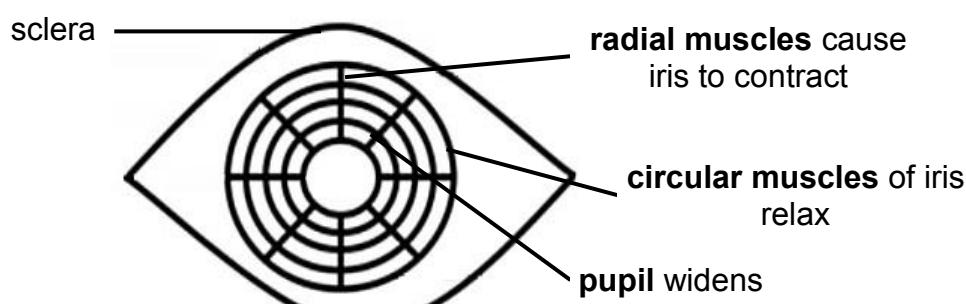


Figure 12B: Dim light conditions

Note: The radial and circular muscles of the iris work antagonistically. This means when one is contracted, the other is relaxed.

Visual defects

1. **Short-sightedness** occurs when a person has the ability to see nearby objects but cannot see distant objects clearly.

Nature of the defect: when looking at distant objects, the light rays focus in front of the retina, causing blurred vision. The defects may be caused by:

- an eyeball that is too long
- the cornea being too curved for the length of the eyeball
- the inability of the lens to become less convex

Treatment: wear glasses with concave lenses

Figures 13A and 13B below show short-sightedness and its treatment.

Defect

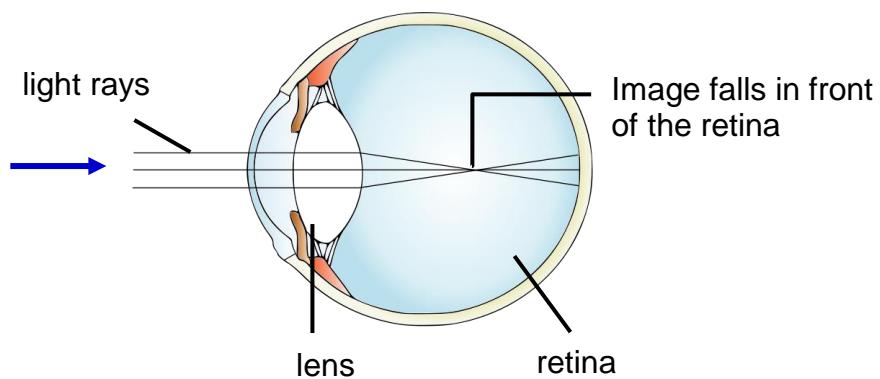
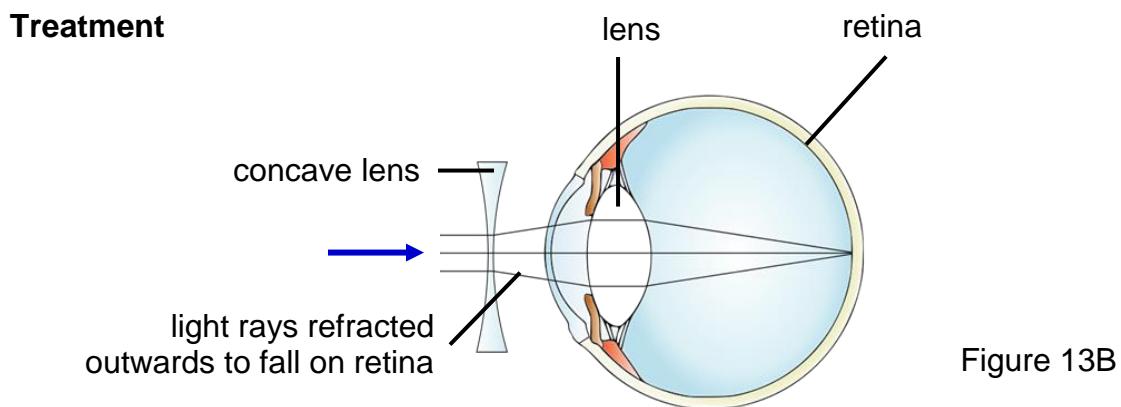


Figure 13A

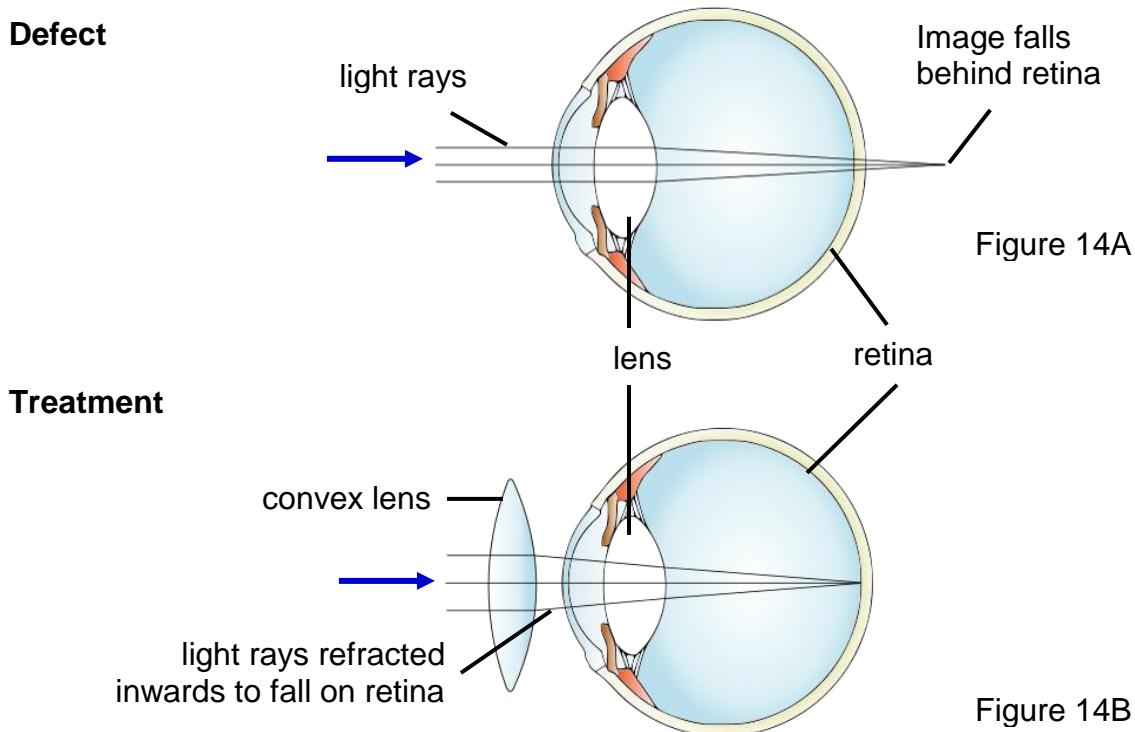


2. **Long-sightedness** occurs when a person has the ability to see distant objects but is unable to see nearby objects clearly.

Nature of the defect: when looking at nearby objects, the light rays focus behind the retina, causing blurred vision. The defects may be caused by:

- an eyeball that is too short (rounded)
- the cornea not being curved enough for the length of the eyeball
- the inability of the lens to become more convex

Treatment: wear glasses with convex lenses



Figures 14A and 14B: Long-sightedness and its treatment

3. **Astigmatism** occurs when the cornea or lens is not equally rounded in all directions as it normally would be (see Figure 15A and 15B).

Normal

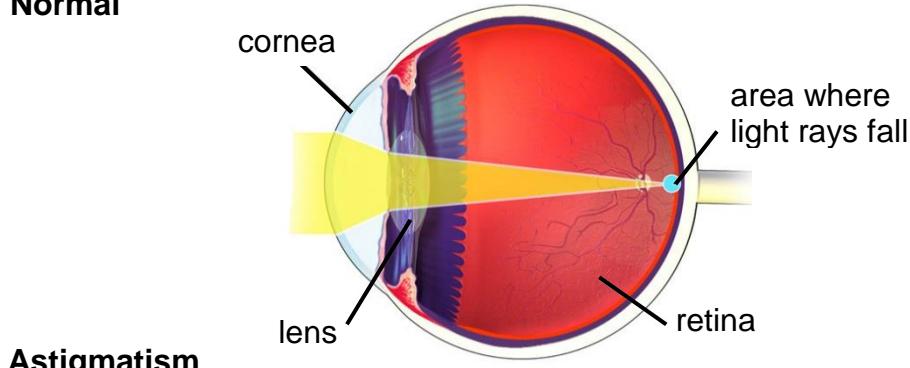


Figure 15A

Astigmatism

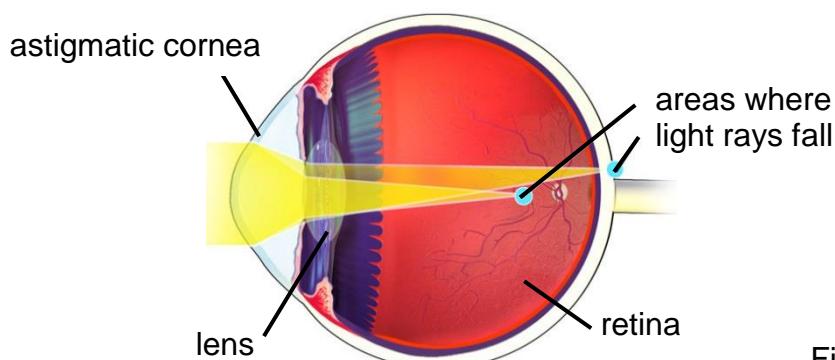


Figure 15B

Figures 15A and 15B: Human eye with normal and astigmatic cornea

With an astigmatic cornea, light entering the eye is not focussed evenly on the retina. This leads to:

- blurred vision
- headaches
- squinting of the eyes

Treatment of astigmatism may include:

- glasses with prescription lenses
- contact lenses
- laser therapy

4. **Cataracts** occur when the clear transparent lens becomes **cloudy**. This prevents light from entering the eye and results in blurred vision. The cataract develops slowly and increases in size over time (see Figures 16A and 16B).

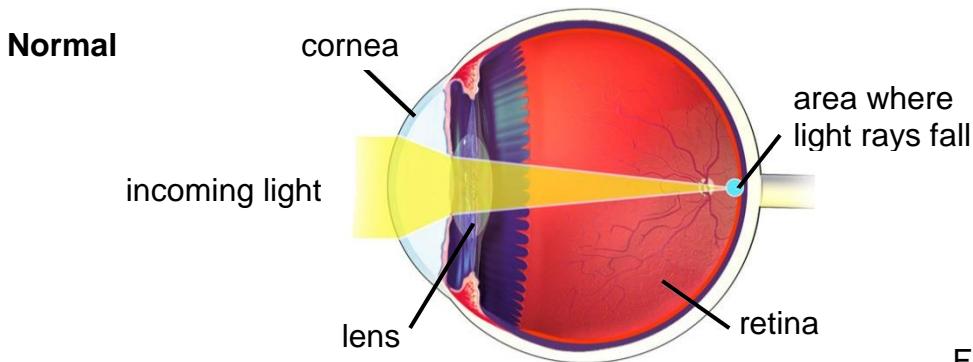


Figure 16A

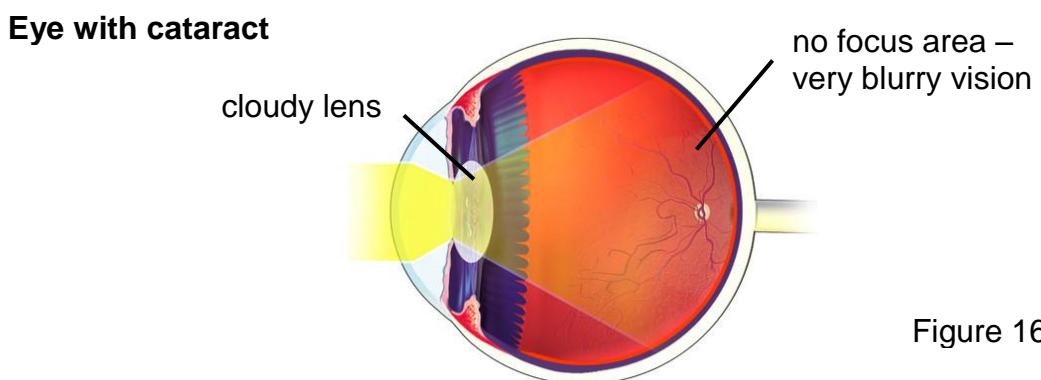


Figure 16B

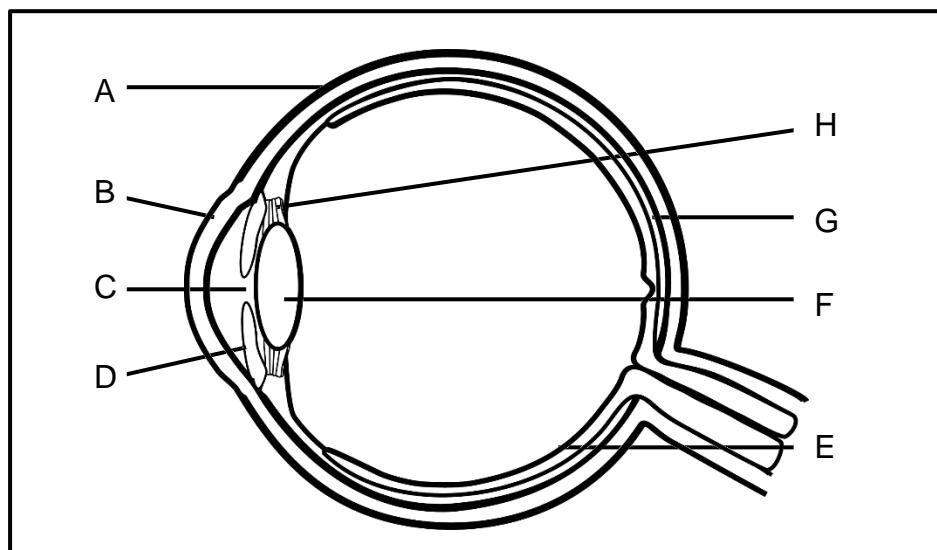
Figures 16A and 16B: Human eye with clear lens and with cataracts

Treatment:

- In the beginning, spectacles may be used.
- As the cataract develops, surgery may be required. During surgery, the lens is removed, and a synthetic lens is inserted.

Activity 5: The human eye

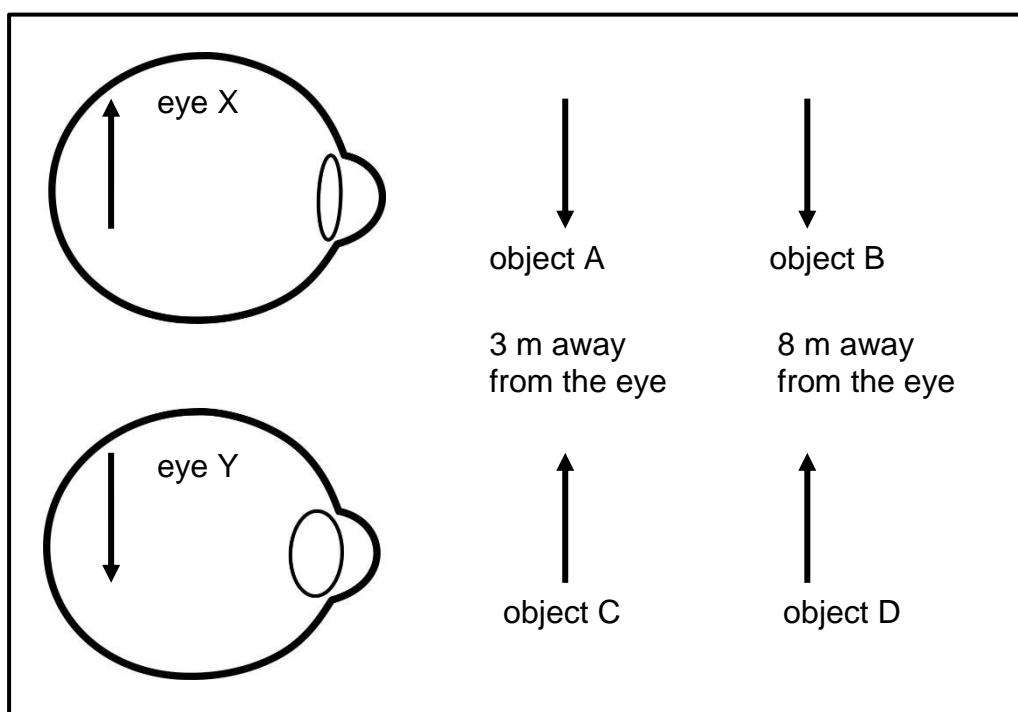
The diagram below represents a section through a human eye.



- Identify parts C, D and H. (3)
 - Explain how part B is adapted to perform its function. (2)
 - Provide the functions of E and G respectively. (2)
 - Write down the letter only for the part that
 - is able to change shape to refract light (1)
 - provides structural support to the eyeball (1)
 - Name and describe the process which involves the iris controlling the amount of light entering the eye when a person is exposed to bright light. (5)
- (14)

Activity 6: Accommodation

The diagram shows two eyes (X and Y) focused on objects (represented by arrows) at different distances from the eye. Objects A and C were 3 metres away from the eye. Objects B and D were 8 metres away from the eye. The diagrams below are not drawn to scale.



- Write down the letter of the object that:
 - eye Y is focussed on (1)
 - eye X is focussed on (1)
- a) Name the eye defect which results in the inability of the eye Y to focus on the object D. (1)
 - Name the type of lens used to rectify the defect in 2(a) above. (1)

3. Identify and describe the process that allows eye Y to form a clear image on the retina. (5)
(9)

Activity 7: Case study on visual defects

Read the extract below and answer the questions that follow.

Kayise, age 60, had failed her vision test for her driver's license. All her life she had suffered from extreme near-sightedness. In fact, without her glasses she was legally blind. When she was denied her license renewal, Kayise came to Dr Nobadula for help. During an examination, it became clear that Kayise had cataracts in both eyes. A cataract is a clouding of the lens in the eye, which reduces vision. The lens is inside the eye and focuses light onto the retina at the back of the eye, where an image is recorded. The lens also adjusts the eye's focus. The lens is made of mainly water and protein. The latter is arranged so the lens stays clear, allowing light to pass through it. Yet, with age, the protein may form clumps that cloud the lens. This is a cataract. The best solution to Kayise's situation was surgery that removed the cataracts and implanted tiny artificial lenses within the eye. Dr Nobadula conducted this procedure. Today, for the first time in her life, Kayise enjoys 20/20 vision without glasses.

1. Describe what is meant by near-sightedness. (2)
 2. Which type of lens can be used to correct this defect? (1)
 3. What causes the clouding to form in the lens? (1)
 4. In your opinion, what would happen to Kayise if she did not have surgery? (1)
 5. Explain the cause of Kayise's near-sightedness and why she did not need glasses after surgery. (4)
 6. Describe what happens to light rays in the eye of a person with cataracts. (3)
 7. Name one other visual defect not mentioned in the extract above. (1)
- (13)

The human ear

Key terminology

organ of Corti	receptor for hearing
crista (plural: cristae)	receptor which detects changes in speed and direction of the head
maculae	receptor which detects changes in the position of the head
semicircular canals	canals which are fluid filled and contain receptors

ampulla (plural: ampullae)	swelling at the base of the semicircular canals which contains the crista
vestibule	structure made up by the sacculus and utriculus, these contain the maculae
grommet	small structure inserted into the tympanic membrane; has a hole through the middle to allow air flow

Figure 17 below shows the structure of the ear.

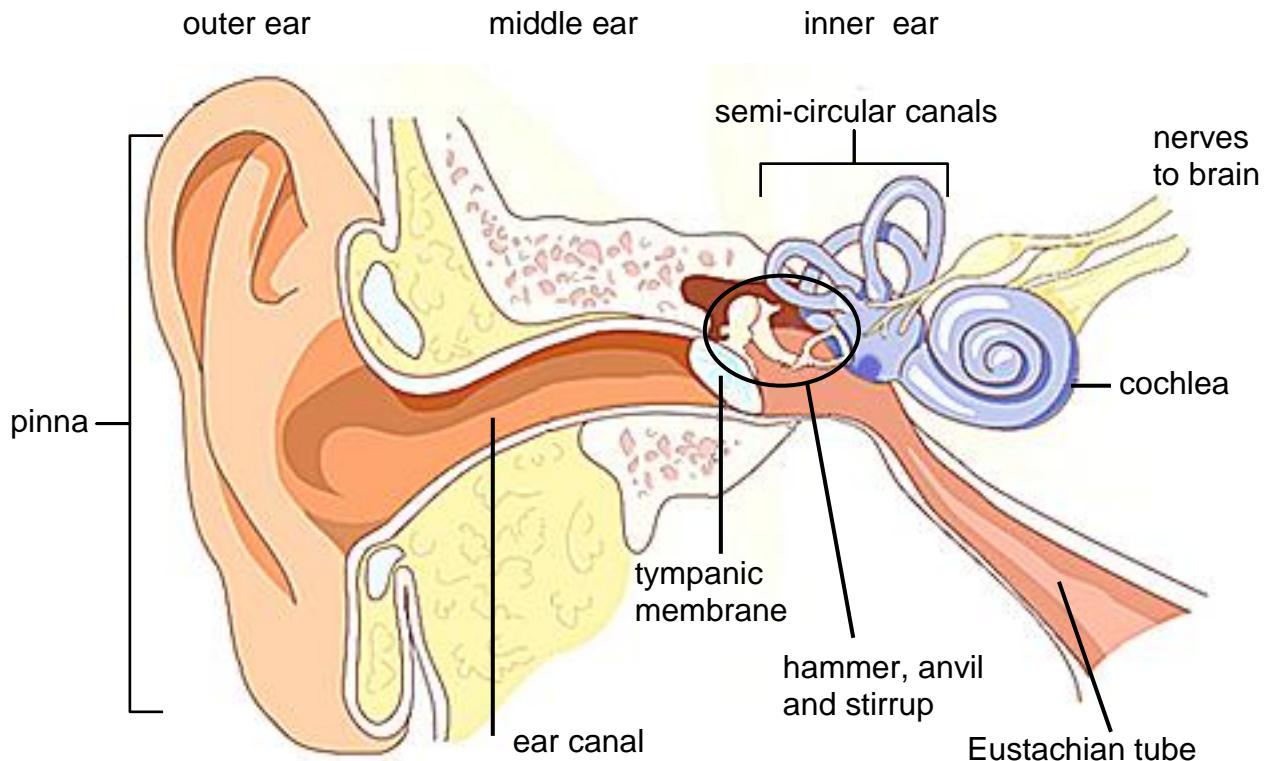


Figure 17: Structure of the human ear

As shown above, the ear may be divided into three sections: the outer ear (Figure 18), the middle ear (Figure 19) and the inner ear (Figure 20).

The outer ear

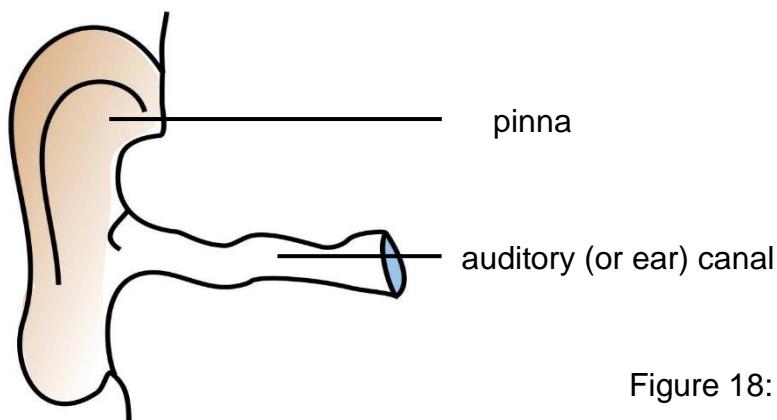


Figure 18: The outer ear

Table 6 below lists the structural adaptations and functions of the various parts of the outer ear.

Table 6: Structural adaptions and function of the outer ear

pinna	<ul style="list-style-type: none"> • cartilage flaps situated on the outside of the ear • direct sound waves into the auditory canal
auditory canal	
<ul style="list-style-type: none"> • tube which passes from the pinna to the tympanic membrane 	<ul style="list-style-type: none"> • transmits sound waves to the tympanic membrane • has little hairs which prevent foreign bodies from entering the ear • has wax which prevents the tympanic membrane from drying out

The middle ear

The middle ear (Figure 19) is an air-filled cavity within the skull. It is separated from the outer ear by the tympanic membrane and is separated from the inner ear by the round window and the oval window. Table 7 list its structural adaptations and functions.

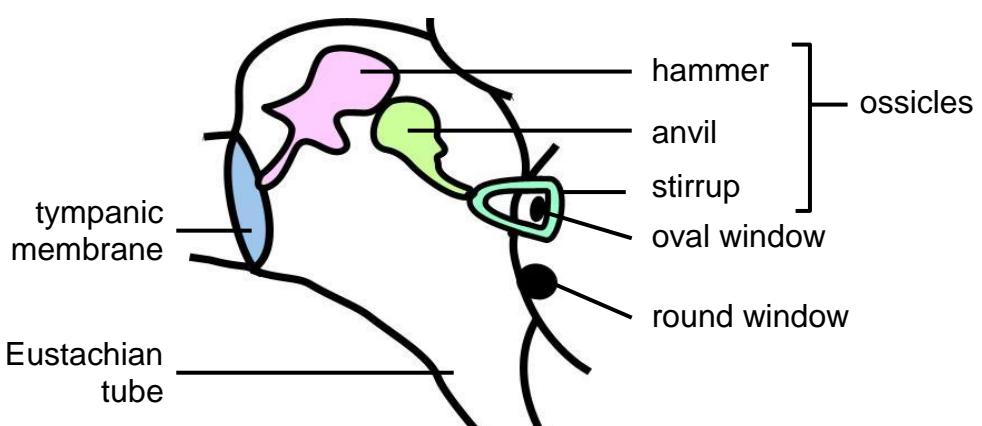


Figure 19: The middle ear

Table 7: Structural adaptions and functions in the middle ear

tympanic membrane	<ul style="list-style-type: none"> • thin membrane separating the inner ear from the middle ear • sound waves moving in auditory canal cause it to vibrate • transmits sound waves to the middle ear
--------------------------	---

ossicles	
<ul style="list-style-type: none"> three irregularly shaped bones: <ul style="list-style-type: none"> hammer (malleus) – largest, connected to tympanic membrane anvil (incus) – middle bone, joining the malleus to the stapes stirrup (stapes) – smallest, connected to the round window 	<ul style="list-style-type: none"> vibrations from the tympanic membrane are transmitted through the ossicles to the inner ear serve to amplify the vibrations (make them larger)
oval window	
<ul style="list-style-type: none"> membrane separating the middle ear from the inner ear 	<ul style="list-style-type: none"> transmits vibrations from the middle ear to the inner ear
round window	
<ul style="list-style-type: none"> membrane situated below the oval window 	<ul style="list-style-type: none"> absorbs excess pressure waves from the inner ear – stop vibrations being echoed
Eustachian tube	
<ul style="list-style-type: none"> thin tube connecting the middle ear to the back of the throat 	<ul style="list-style-type: none"> equalising pressure on both sides of the tympanic membrane

The inner ear

The inner ear lies within the bones of the skull and is made up of the semi-circular canals, vestibule (sacculus and utriculus) and the cochlear which are continuous with each other (Figure 20). The structural adaptations and functions of the various parts are listed in Table 8.

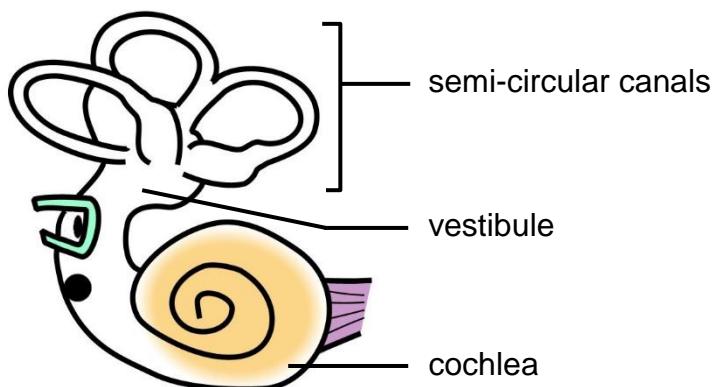
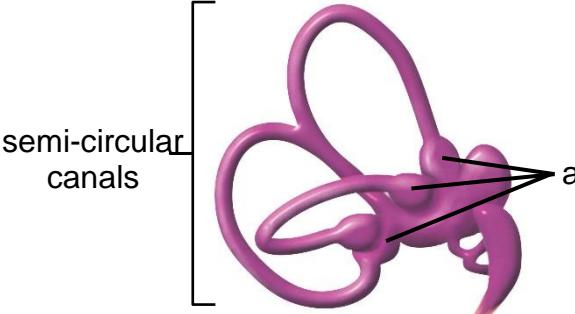
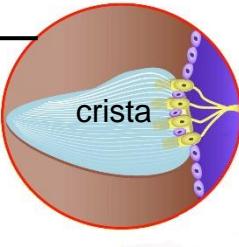
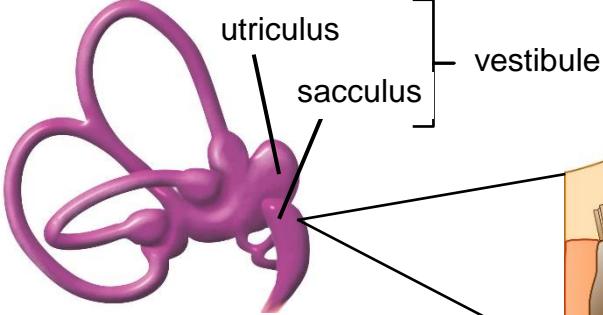
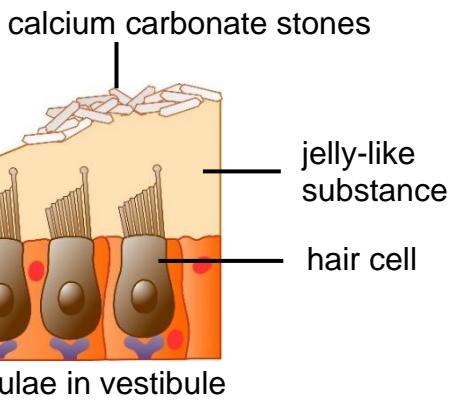


Figure 20: Inner ear

These structures are made up of bony cavities and are called the bony labyrinth. This labyrinth is filled with a fluid called perilymph. Suspended in the perilymph is a

system of membranes called the membranous labyrinth. This labyrinth contains a fluid called endolymph.

Table 8: Structural adaptation and functions of parts of the inner ear

Structural adaptations	Functions
semi-circular canals	
<ul style="list-style-type: none"> three semi-circular canals are arranged at right angles to each other they are located above the vestibule each canal has an enlarged area, called the ampulla, at one end inside the ampulla are receptors (cristae) 	<ul style="list-style-type: none"> cristae detect changes in speed and direction and generate impulses sent to the cerebellum
	
Figure 21: Semi-circular canals with crista in ampulla	
vestibule	
<ul style="list-style-type: none"> made up of two membranous sacs called the sacculus and utriculus (both filled with endolymph); inside each are receptors called maculae maculae have tiny hair cells covered with a jelly-like substance and tiny calcium carbonate stones. 	<ul style="list-style-type: none"> receptors (maculae) detect changes in the position of the head with respect to gravity
	
Figure 22: Vestibule with detail of maculae	

cochlea

- | | |
|---|---|
| <ul style="list-style-type: none">• divided into three chambers• the upper and lower chambers are filled with perilymph• middle chamber is filled with endolymph, and contains the organ of Corti• organ of Corti contains tiny hairs embedded into a membrane | <ul style="list-style-type: none">• organ of Corti is receptor responsible for interpreting sound• it converts the stimulus of sound into an impulse |
|---|---|

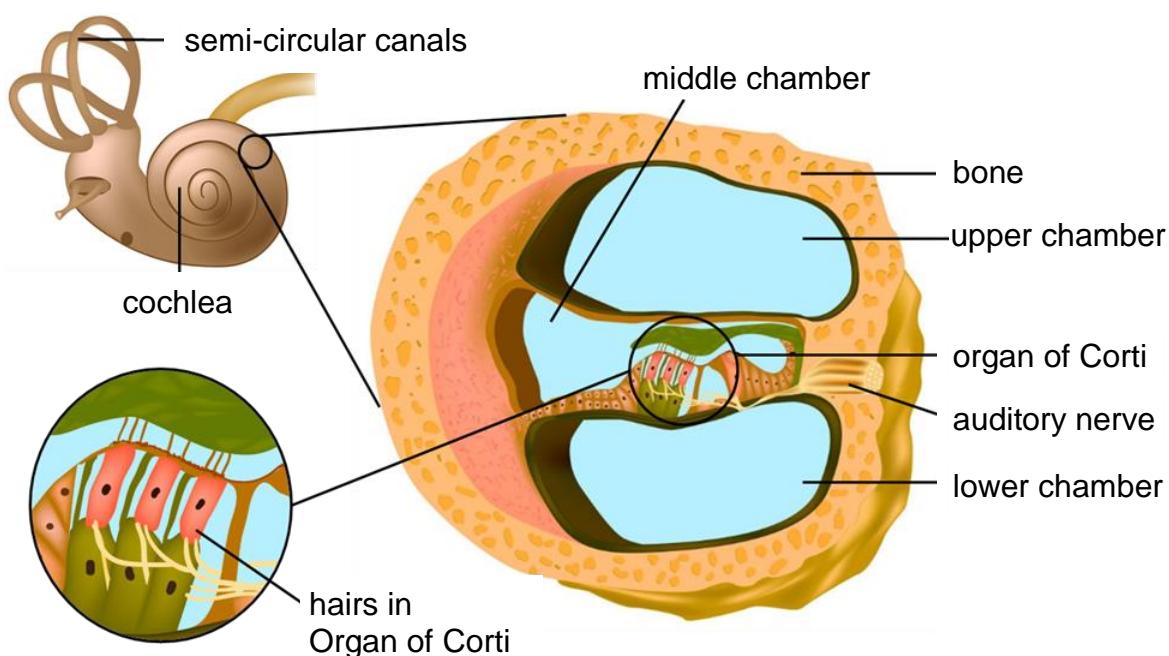
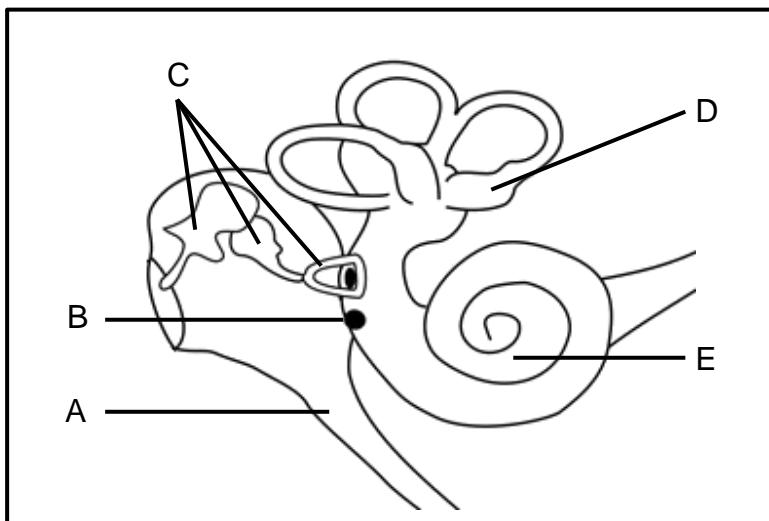


Figure 23: Cochlea, in cross-section, and detail of hairs in the organ of Corti

Structures of the ear: <https://www.youtube.com/watch?v=HMXoHKwWmU8>

Activity 8: The human ear

The diagram below represents a part of the human ear.



1. Identify:
 - a) part A (1)
 - b) part B (1)
 - c) part E (1)
2. Provide the collective name for the bones found at C. State two functions of these bones. (3)
3. The structure labelled D contains receptors.
 - a) Name the receptors found in this structure. (1)
 - b) Give the stimulus to which these receptors respond. (2)(9)

Functioning of the human ear

The human ear has two functions:

- hearing
- maintaining balance

Hearing

Hearing (illustrated in Figure 24 below) is the process in which sound waves are transmitted through the ear and impulses are generated which are sent to the cerebrum for interpretation. This process occurs as follows:

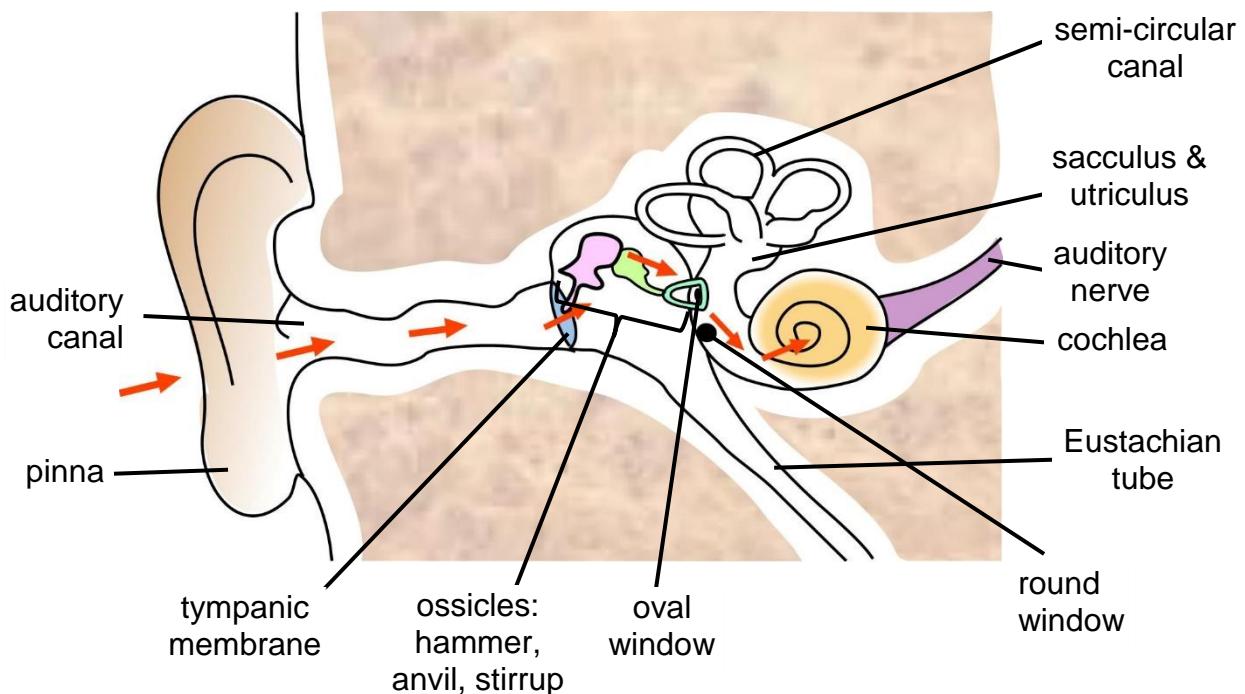


Figure 24: The process of hearing

- The pinna traps and directs sound waves into the auditory canal towards the tympanic membrane.
- The tympanic membrane vibrates as the sound waves strike against it.
- The vibrating tympanic membrane causes the ossicles to vibrate.
- The hammer, anvil and stirrup amplify and transmit the vibrations to the oval window.
- The oval window is smaller than the tympanic membrane. As a result, the pressure increases, causing the sound to be amplified.
- The vibrating oval window causes pressure waves to travel through the endolymph in the cochlea.
- The organ of Corti in the middle chamber of the cochlea is stimulated.
- The stimulus is converted into a nerve impulse which is transmitted to the auditory nerve.
- The auditory nerve transmits the impulse to the cerebrum for interpretation.
- The pressure waves in the cochlea are absorbed into the middle ear through the round window and exit the body via the Eustachian tube.

Maintaining balance

Balance is the process in which receptors in the inner ear detect changes in the position of the head and respond to gravity as well as changes in speed and

- 2.1.6 Suppose the lamp was moved from position 7 to position 2. Describe the mechanism that caused the change in the diameter of the pupil. (4)
- 2.1.7 Name the process mentioned in question 2.1.6. (1)
- 2.1.8 Plot a bar graph to represent the data gathered during this investigation. (7)
- (21)
- 2.2 Read the extract below and answer the questions that follow.

A LINK BETWEEN CONCUSSION AND BRAIN DAMAGE

In 2002 a former American football player was found dead in his truck. The doctor who handled the autopsy discovered that the football player had severe brain damage and that his death was caused by repeated blows to the head or repeated concussions. He called this disorder chronic traumatic encephalopathy (CTE).

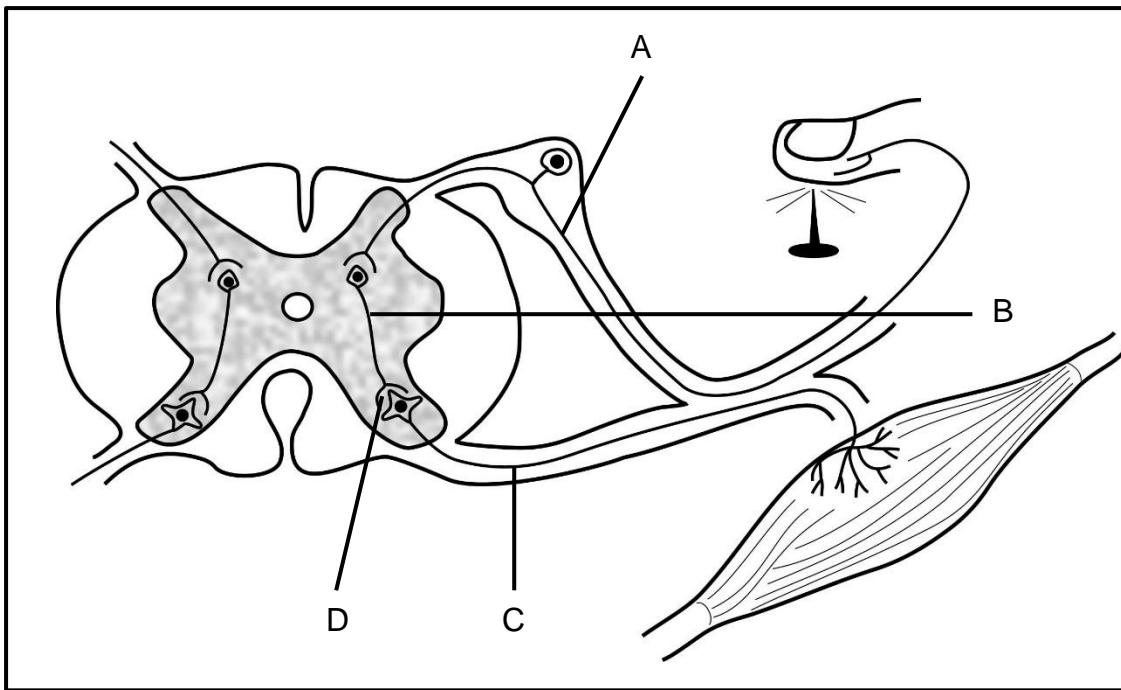
A more recent study was conducted that involved the brains of 165 people who played football at high school, college or professional level. The study found evidence of CTE in 131 of the brains.

(adapted from www.wikipedia.org and www.theatlantic.com)

- 2.2.1 The part of the brain affected by CTE is the cerebrum.
State two possible symptoms of this disorder. (2)
- 2.2.2 State one way in which the brain is protected. (1)
- 2.2.3 Explain why CTE does not usually affect essential life processes such as breathing or heart rate. (2)
- (5)
- [26]

Question 3

- 3.1 Study the diagram of a reflex arc below.



- 3.1.1 What is a reflex action? (1)
- 3.1.2 Label the following:
- The functional connection at **D**. (1)
 - Neuron **B** (1)
- 3.1.3 State the significance of the microscopic gap indicated by **D** (1)
- 3.1.4 Write down in the correct order, the letters only of the neurons involved from the time a stimulus is received until a response takes place. (2)
- 3.1.5 Explain the consequences for a reflex action if neuron **C** is damaged. (2)
- 3.1.6 Draw a labelled diagram to represent the structure of neuron **A**. (5)
(13)
- 3.2 Read the article below and answer the questions that follow.

The discovery of Alzheimer's Disease (AD)

Alzheimer's disease (AD) is an irreversible brain disease that slowly destroys brain cells, causing loss in memory and thinking skills serious enough to interfere with daily life. The symptoms first appear after the age of 60, making it the most common cause of dementia among older people. (continued on next page ..)

Continued: It is a neurological brain disorder named after a German physician, Alois Alzheimer, who first described it in 1906. He noticed changes in the brain of a woman who had died after an unusual mental illness. Her symptoms included memory loss, language problems and strange behaviour. After she died he inspected her brain and found many clumps and tangled bundles of nerve fibres.

Abnormal clumps, tangled-bundles of nerve fibres and the loss of connections between brain cells are all symptoms of the disease. AD gets worse over time, with death due to organ failure usually occurring two to eight years after the start. At present there is no cure.

adapted from: www.ALZinfo.org. Fischer Centre for Alzheimer's Research Foundation.

The table below presented by the World Health Organisation shows the percentage of people in the general western population affected by AD in different age groups.

Age groups (years)	Percentage patients with AD (%)
65-69	1,4
70-74	2,8
75-79	6,6
80-84	11,1
85+	23,6

- 3.2.1 What was the percentage increase of patients with AD, between the oldest two age groups? (1)
- 3.2.2 What seems to be the earliest symptoms of Alzheimer's disease? (1)
- 3.2.3 Describe what the person's brain looked like when it was dissected. (2)
- 3.2.4 What is the main cause of death in a person with AD? (1)
- 3.2.5 Use the table to plot a histogram to show the occurrence of AD. (6) (11)
[24]

Section B: [50]

Total marks: [90]

7: The human endocrine system and homeostasis

Introduction

The difference between the important secretory glands

Endocrine glands

The hypothalamus and pituitary gland

Adrenal glands

Reproductive glands

Activity 1: Endocrine glands and their hormones

Activity 2: Effects of a hormone

Negative feedback

Homeostatic control of the internal environment

The main mechanisms used by the body to maintain homeostasis

Negative feedback in the regulation (control) of water balance – osmoregulation

Negative feedback in the regulation of salt concentration

Negative feedback in the regulation of carbon dioxide (CO_2) concentrations

Negative feedback mechanism in the regulation of blood glucose levels

Negative feedback in the regulation of thyroxin levels

Activity 3: A negative feedback mechanism

Endocrine system disorders

Pituitary gland disorders

Thyroid gland disorders

Pancreas disorders

Activity 4: Research task endocrine disorders

Regulation of body temperature – thermoregulation

Activity 5: Body temperature

End of topic exercises

CHAPTER 7: THE HUMAN ENDOCRINE SYSTEM AND HOMEOSTASIS

Introduction

Various human mechanisms enable us to respond and react to the outside environment so as to maintain a constant internal environment. Our responses are controlled by the **nervous** and **endocrine** systems and to an extent, the **immune** system. The working together of these systems helps to maintain stability within the organism and so protects us.

The human nervous system responds via:

- a rapid response to stimuli
- the use of electrical impulses and neurotransmitters

The endocrine response is controlled by the endocrine glands situated throughout the body. This response is:

- a slower response that has a long-lived effect
- as the endocrine glands produce and release specific hormones into the bloodstream.
- An effector organ is targeted, and
- a response is initiated.
- When endocrine organs are either over- or under stimulated, endocrine disorders are observed.

Key terminology

endocrine system	a system responsible for chemical co-ordination and regulation of various activities in the body
homeostasis	a process of maintaining a constant internal environment (blood and tissue fluid) within the body.
hormones	chemical messengers in the body. they travel in the bloodstream and cause an effect elsewhere in the body
negative feedback	operate in the human body to detect changes or imbalances in the internal environment and to restore balance

osmoregulation	regulation of the water balance in the internal environment
osmotic pressure	a measure of the concentration of solutes (e.g. salt, glucose) present in a solution; this may determine whether a cell loses or gains water
antagonistically	to work in opposite ways; if one hormone causes an increase of a substance, the other hormone will cause a decrease of that substance, e.g. insulin and glucagon
thermoregulation	the control of the body temperature to keep it as close to 37°C as possible
endothermic	relates to an organism that generates heat internally through a metabolic process to maintain a constant body temperature
vasoconstriction	narrowing of blood vessels
vasodilation	widening of blood vessels
evaporation	heat loss when sweat changes into water vapour on the surface of the skin
conduction	transfer of heat between objects which are in contact
convection	as warm air rises it is replaced by cooler air
radiation	heat transfer between two objects which are not in contact

Homeostasis is the tendency of an organism or cell to regulate its internal conditions, usually by a system of feedback controls, so as to stabilize health and functioning, regardless of the outside changing conditions.

The difference between the important secretory glands

Mammals produce secretions from exocrine and endocrine glands. The main differences between the two are listed in Table 1 below. Examples of each are given together with their secretions.

Table 1: Exocrine and endocrine glands – the main differences

Exocrine glands	Endocrine glands
<ul style="list-style-type: none"> • have ducts • secretions released into a cavity or on a surface • <i>Examples:</i> salivary glands (saliva), sweat glands(sweat) 	<ul style="list-style-type: none"> • ductless • hormones released into the bloodstream • <i>Examples:</i> pituitary (ADH), thyroid (TSH), pancreas (insulin)

Note: The pancreas is both an endocrine and an exocrine gland. As an exocrine

gland it secretes digestive enzymes into the small intestine. As an endocrine gland it secretes insulin and glucagon into the bloodstream.

Figures 1 and 2 give examples of exocrine and endocrine glands with their secretions.

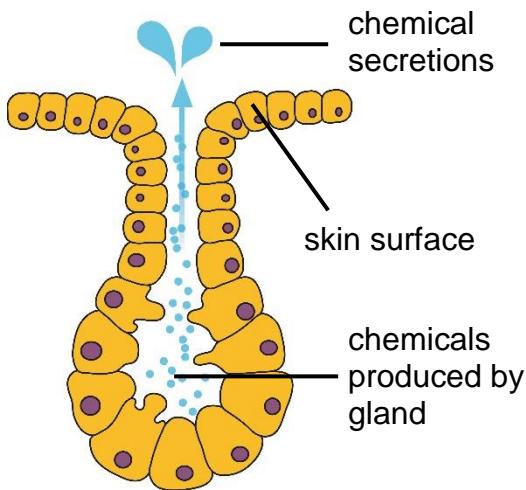


Figure 1: Exocrine gland

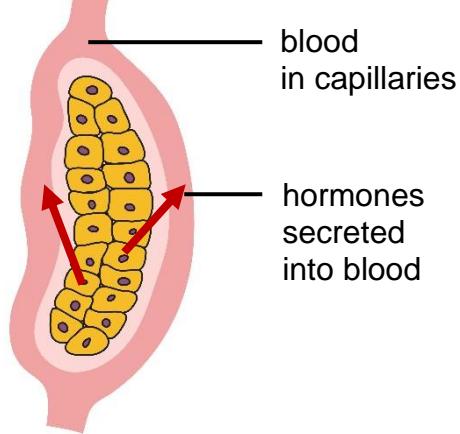


Figure 2: Endocrine gland

Endocrine glands

The endocrine system has many important endocrine glands that secrete hormones into the bloodstream where they are transported to their target site (such as an organ). These hormones do not work in isolation and often interact with other hormones in a sequence of events. They can work to produce a common effect, or they can work antagonistically.

Hormones: Hormones are organic compounds that act as messengers in the body. Most hormones are proteins with some being steroids (lipids). They are only needed in small amounts and they give a more lasting response than a nerve response. Hormones may be over-secreted or under secreted resulting in certain disorders.

The basic functions of hormones are:

- reproduction, growth and development
- maintenance of the internal environment (by **stimulating** or **inhibiting** the functioning of cells / organ)
- regulation of metabolism by controlling production, usage and storage of energy

Figure 3 shows the location of the human endocrine glands and the hormones produced there. The function of the hormones is discussed later.

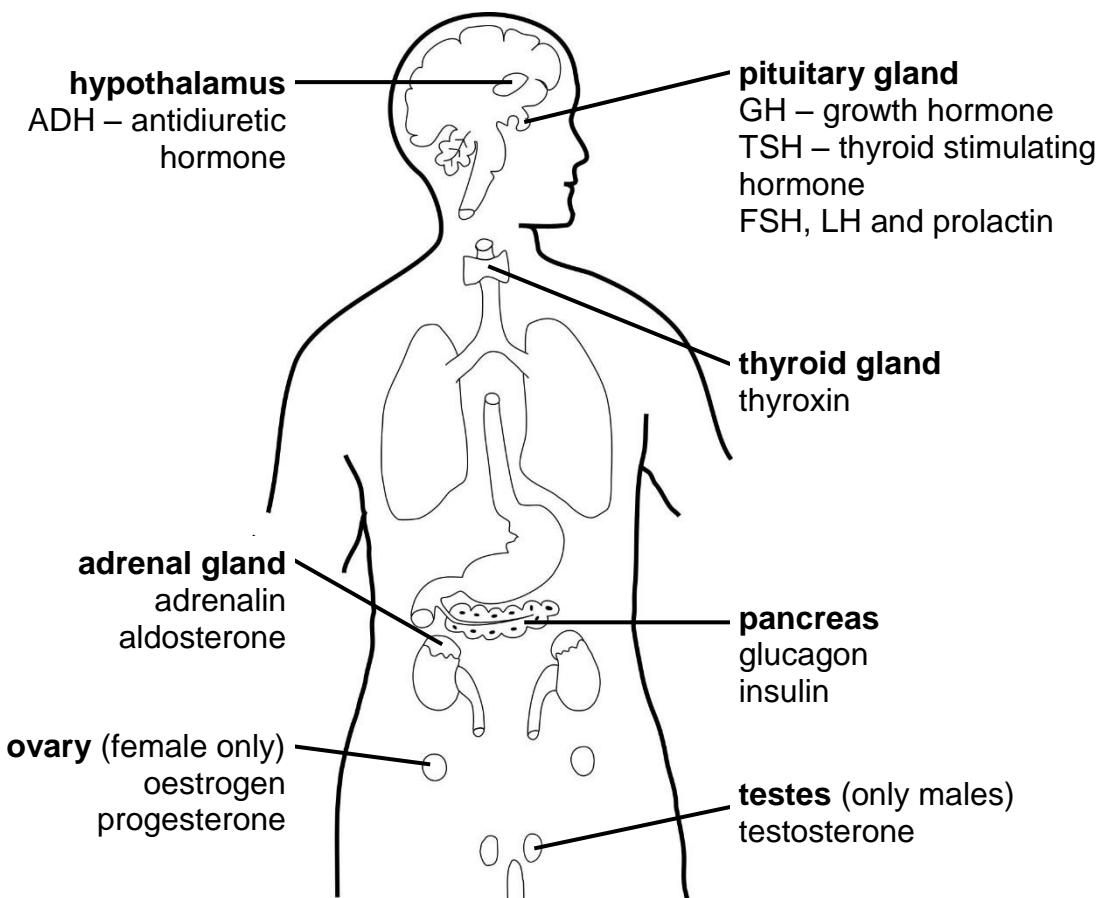


Figure 3: Endocrine glands and the hormones they secrete

Introduction to the endocrine system:

<https://www.youtube.com/watch?v=SHgNaomqlRA>

The hypothalamus and pituitary gland

The hypothalamus is a small area of the human brain, located above and connected to the pituitary gland (see Figure 4). It produces and secretes important hormones and is a link between the nervous and the endocrine systems.

The hypothalamus plays an important role in controlling vital functions in the human body. In this text the focus is on the anti-diuretic hormone (ADH), produced by the hypothalamus.

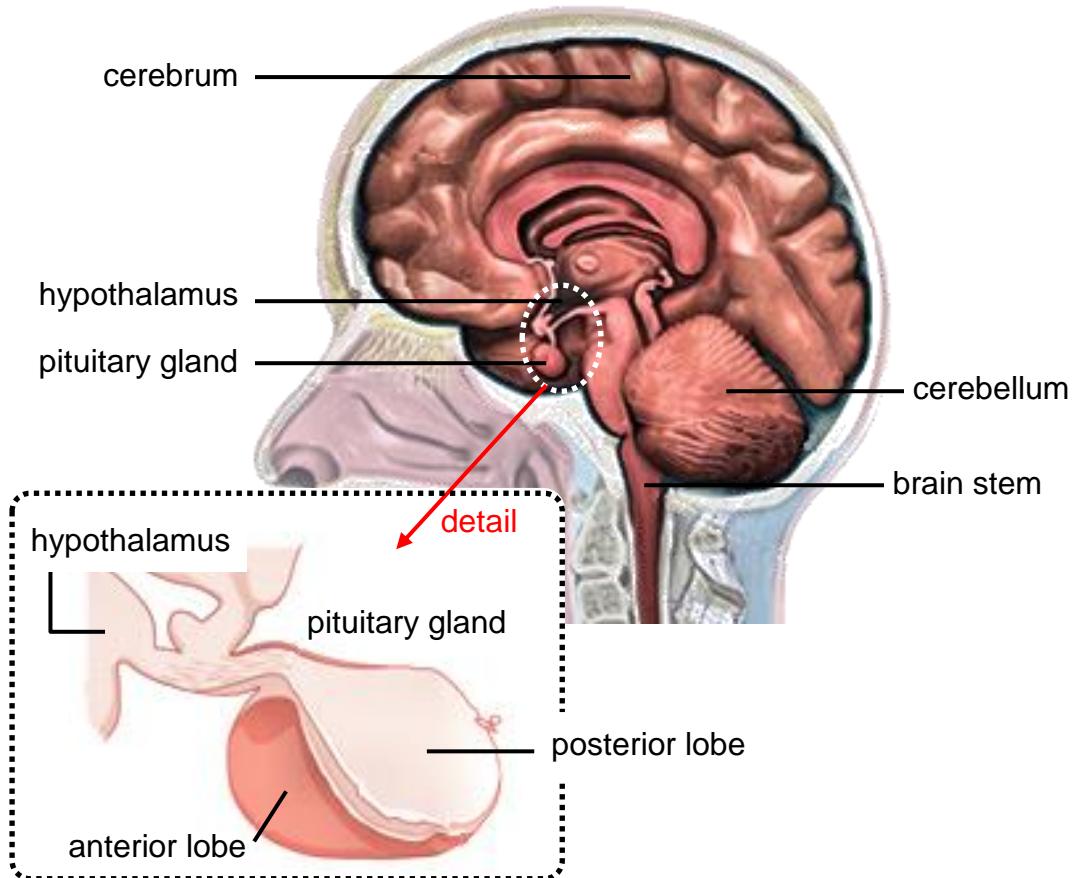
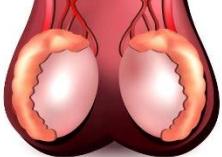
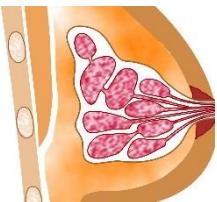


Figure 4: Location of hypothalamus and pituitary gland in the human brain, with detail of link between the two glands.

Table 2 below lists the hormones released by the hypothalamus and the pituitary gland. The function and target organ/s or gland/s are also given.

Table 2: Endocrine glands, the hormones secreted and their function and target

Hormones secreted	Target organ / gland	Functions of hormone
hypothalamus		
<ul style="list-style-type: none"> • ADH (antidiuretic hormone), also called vasopressin 	<ul style="list-style-type: none"> • kidneys 	<ul style="list-style-type: none"> • stimulates the re-absorption of H₂O into the tubules • protects the body against dehydration
pituitary gland (anterior lobe)		
<ul style="list-style-type: none"> • TSH (thyroid stimulating hormone) 	<ul style="list-style-type: none"> • thyroid gland 	<ul style="list-style-type: none"> • stimulates growth of thyroid gland • stimulates thyroid gland to secrete the hormone thyroxin

<ul style="list-style-type: none"> • FSH (follicle stimulating hormone) 	<ul style="list-style-type: none"> • ovaries 	<ul style="list-style-type: none"> • stimulates development of follicles • stimulates the ovaries to produce the hormone oestrogen • stimulates the development of ova (eggs) in the female
	<ul style="list-style-type: none"> • testes 	<ul style="list-style-type: none"> • stimulates the testes to produce spermatozoa
<ul style="list-style-type: none"> • LH (luteinising hormone) 	<ul style="list-style-type: none"> • ovaries 	<ul style="list-style-type: none"> • stimulates ova maturation • stimulates ovulation (release of the ovum)
	<ul style="list-style-type: none"> • testes 	<ul style="list-style-type: none"> • stimulates the production of the male hormone testosterone
<ul style="list-style-type: none"> • prolactin 	<ul style="list-style-type: none"> • mammary glands 	<ul style="list-style-type: none"> • stimulates milk production and secretion
<ul style="list-style-type: none"> • GH (growth hormone) 	<ul style="list-style-type: none"> • Bone and muscle cells 	<ul style="list-style-type: none"> • Stimulates the growth of long bones and skeletal muscles

Other important endocrine glands that make up the endocrine system are:

- the adrenal glands,
- the ovary in females and the testes in males,
- the pancreas, and
- the thyroid gland.

All the endocrine glands are essential for ensuring healthy metabolism.

Adrenal glands

These two triangular shaped glands are situated on top of each of the kidneys (see Figure 5). The adrenal glands have a central medulla region and an outer cortex. The two hormones produced and secreted from the adrenals, that we will discuss, are **adrenalin** and **aldosterone** (see Table 3).

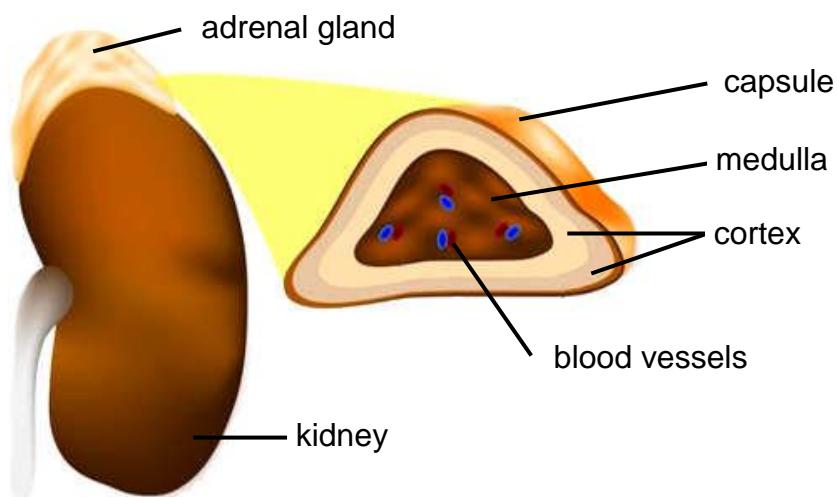


Figure 5: Location and structure of one of the adrenal glands

Table 3: Adrenal hormones, target organs / areas and their functions

Target areas / organs	Functions
adrenalin (the 'fight or flight' hormone)	
<ul style="list-style-type: none">• heart• liver• skeletal muscles• eye muscles• skin• lungs• body cells	<ul style="list-style-type: none">• increased heart rate and blood supply to cardiac muscles• stimulates the liver to increase conversion of glycogen into glucose• increased blood supply to skeletal muscle• stimulates pupil dilation• decreased blood supply to 'less vital' organs (digestive system and the skin)• increased breathing rate• increased metabolic rate
aldosterone	
<ul style="list-style-type: none">• kidneys	<ul style="list-style-type: none">• regulates the salt (sodium / Na^+ and potassium / K^+) concentration in the blood; works together with ADH to achieve this

The reproductive glands

The hormones released by the human reproductive glands (see Figures 6A & 6B) are important in reproduction and for stimulating the secondary characteristics at onset of puberty.

In females the ovaries are stimulated by FSH from the pituitary gland. The ovaries themselves release oestrogen and progesterone which play different roles in the menstrual cycle. In males the cells of Leydig in the testes secrete the male hormone testosterone which stimulates sperm production and maturation.

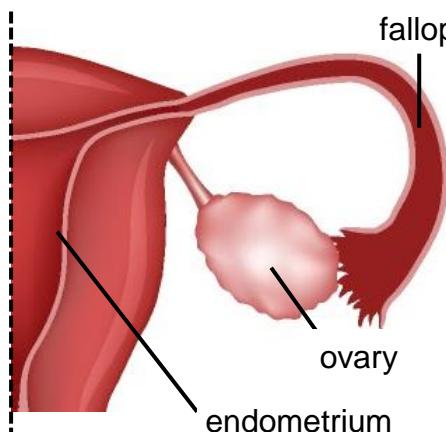


Figure 6A: Ovary

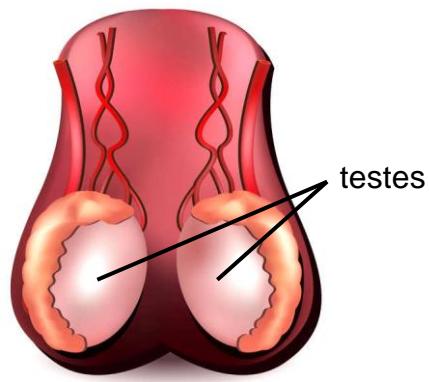


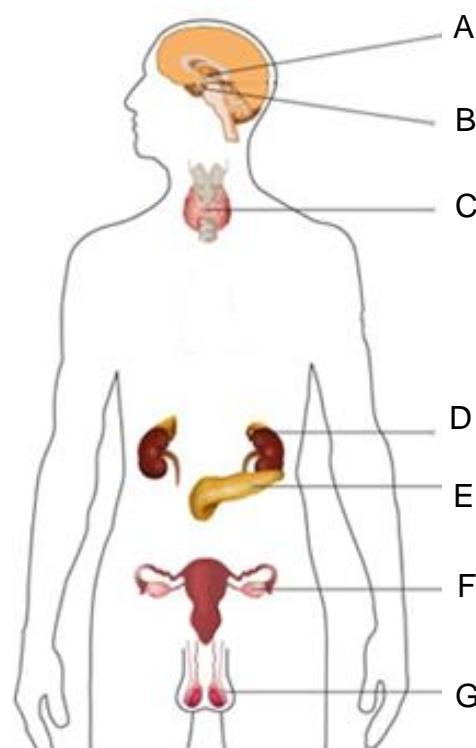
Figure 6B: Testes

Table 4: Hormones released by the human reproductive glands and their functions

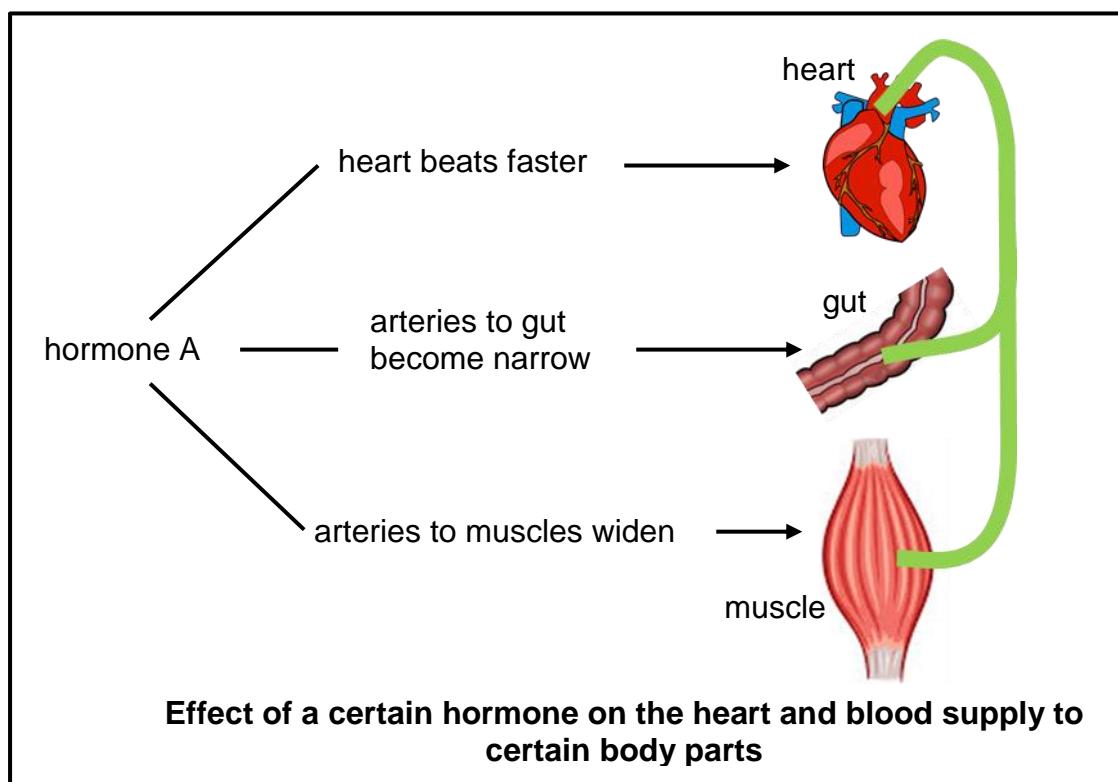
Ovaries	Testes
<p>Oestrogen</p> <ul style="list-style-type: none">• promotes the thickening of the endometrial wall• promotes the development of the female secondary sexual characteristics at puberty• promotes fertility <p>Progesterone</p> <ul style="list-style-type: none">• promotes further thickening and vascularisation of the endometrial wall• maintains implantation of embryo during pregnancy	<p>Testosterone</p> <ul style="list-style-type: none">• stimulates the development of the male sex organs and the secondary characteristics during puberty• promotes the maturation of the sperm

Activity 1: Endocrine glands and their hormones

The diagram shows some of the human endocrine glands. Name the glands labelled A to G, and list the hormone/s produced by each gland. (20)



Activity 2: Effects of a hormone



1. Give the name of hormone A. (1)
 2. State the position of the gland that secretes hormone A in the human body. (1)
 3. Explain the importance of the narrowing of the arteries to the gut under emergency conditions. (4)
 4. Name the part of the human eye that is also affected by hormone A. (1)
 5. Explain the influence of hormone A on the part named in question 4. (3)
- (10)

Negative Feedback

A negative feedback mechanism is an interaction between two hormones in which one hormone stimulates an increase in another hormone which then inhibits the first hormone, thus restoring balance.

The following is the general sequence of events in a negative feedback mechanism:

- an imbalance is detected by the receptor
- a control centre is **stimulated**
- the control centre **responds**
- a **message** sent to target organ/s which are the effectors
- the effector **responds**
- it **opposes (reverses)** the imbalance
- **balance** is restored

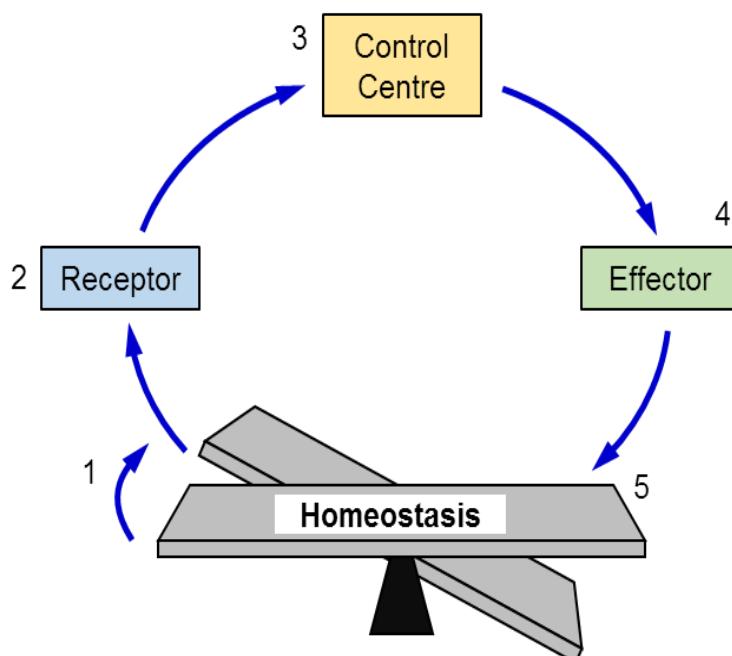


Figure 7: Homeostasis mechanism

As illustrated in Figure 7, an imbalance triggers a stimulus to the receptors at 2. When stimulated, the receptors generate an impulse that is transmitted to the control centre at 3. The control centre responds by sending an impulse to the effector which is the target organ at 4. The target organ then reverses the imbalance and balance is restored at 5.

Homeostatic control of the internal environment

The tissue fluid which surrounds our cells constitute the internal environment. The conditions within cells depend on the conditions within the internal environment. When faced with changes from either the external or internal environment the human body controls this effect and **homeostasis** is achieved.

Without homeostasis, organs, systems and ultimately, the whole organism, may be negatively affected. The important variables and homeostatic mechanisms that will be discussed are the:

- maintenance of water levels
 - maintenance of salt levels
 - regulation of thyroxin levels
-
- maintenance of glucose levels
 - regulation of the CO₂ concentrations
 - regulation of body temperature

It is important to understand the reasons why certain factors must be controlled and how this is carried out.

The main mechanisms for maintaining homeostasis

Table 5 below is a summary of six important homeostatic controls humans possess to ensure stability within their internal environment.

Table 5: Homeostatic control of internal environment

Stimulus	Reason for importance of regulation	Effector/s
Water	All metabolic reactions require a balance of water and salt concentrations in the blood and surrounding tissue fluid.	Kidneys and the skin
Salts	The presence of dissolved salts in the blood and tissue fluid determines the osmotic pressure which may contribute to the cell losing or gaining water by osmosis affecting the balance of these fluids	Kidneys

CO₂	The presence of CO ₂ produced during cellular respiration affects the pH of the blood and tissue fluid. Enzymes are extremely sensitive to pH variations.	Lungs
Glucose	The concentration of glucose in the body needs to be controlled to ensure that energy levels can be maintained, and metabolic functioning can continue.	Liver and pancreas
Thyroxin (T4)	Cellular metabolism is regulated by the hormone thyroxin. Slight variations in the amount of thyroxin can have a severe effect on the metabolic rate of an individual.	Thyroid gland
Temperature	An increase or decrease from normal body temperature can affect metabolism due to the effect on enzyme activity. High temperatures denature enzymes whereas low temperatures slow down enzyme activity.	Skin

Negative feedback in the control of water balance – OSMOREGULATION

The maintenance of the water balance is very important to ensure that body metabolism continues. The homeostatic control of water and salt levels in blood and tissue fluid is carried out in a negative feedback system known as **osmoregulation**.

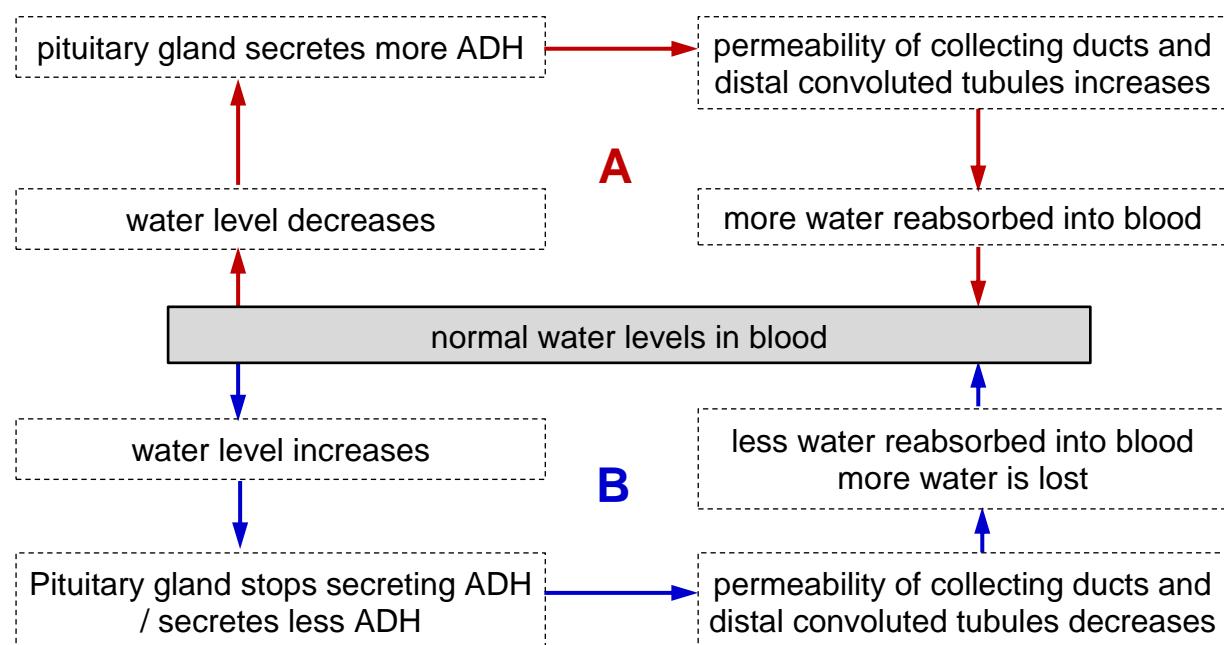


Figure 8: Negative feedback control of water levels in blood and tissue fluid

Figure 8 above and Table 6 below show how the body regulates:

- A Levels of water when there is a drop **below normal** (dehydration), and
- B Levels of water when there is an increase **above normal** (overhydration)

Table 6: Osmoregulation

A – decreased water level	B – increased water level
Dehydration - when the water levels in the blood and tissue fluids are low	Overhydration is when water levels in the blood and tissue fluids are high.
May be due to excessive exercise, hot temperatures, increased sweating or decreased water intake.	May be due to cooler temperatures, little exercise with no sweating and an excessive intake of water.
Low levels of water in the blood and tissue fluid is detected by receptor cells (osmoreceptors), in the hypothalamus of the brain.	Water levels in the blood and tissue fluid are high, and this is detected by the osmoreceptors in the hypothalamus .
Impulses are sent to the pituitary gland and antidiuretic hormone (ADH) is released.	Impulses are sent to the pituitary gland and less anti-diuretic hormone (ADH) is released.
The hormone is transported in the blood to the effector organ , the kidney . The permeability of the collecting duct and the distal convoluted tubule is increased .	Collecting ducts and distal convoluted tubules in the kidney become less permeable .
More water is reabsorbed and passed into the blood .	Less water is reabsorbed into the blood .
The blood becomes more dilute and concentrated urine is excreted .	More water is lost, more dilute urine is excreted .

Water levels in the blood and tissue fluid return to normal and **homeostasis** is achieved.

Negative feedback in the regulation of salt concentration

The osmotic pressure in the blood and tissue fluids is affected by the presence of solutes. Glucose and salts make up the solutes. **Sodium ions (Na⁺) and potassium ions (K⁺)** are salts that are regulated in a negative feedback system.

Figure 9 and Table 7 show how the body regulates the:

- A** levels of salt concentration in the blood when they drop below normal, and
B levels of salt concentration in the blood when they rise above normal.

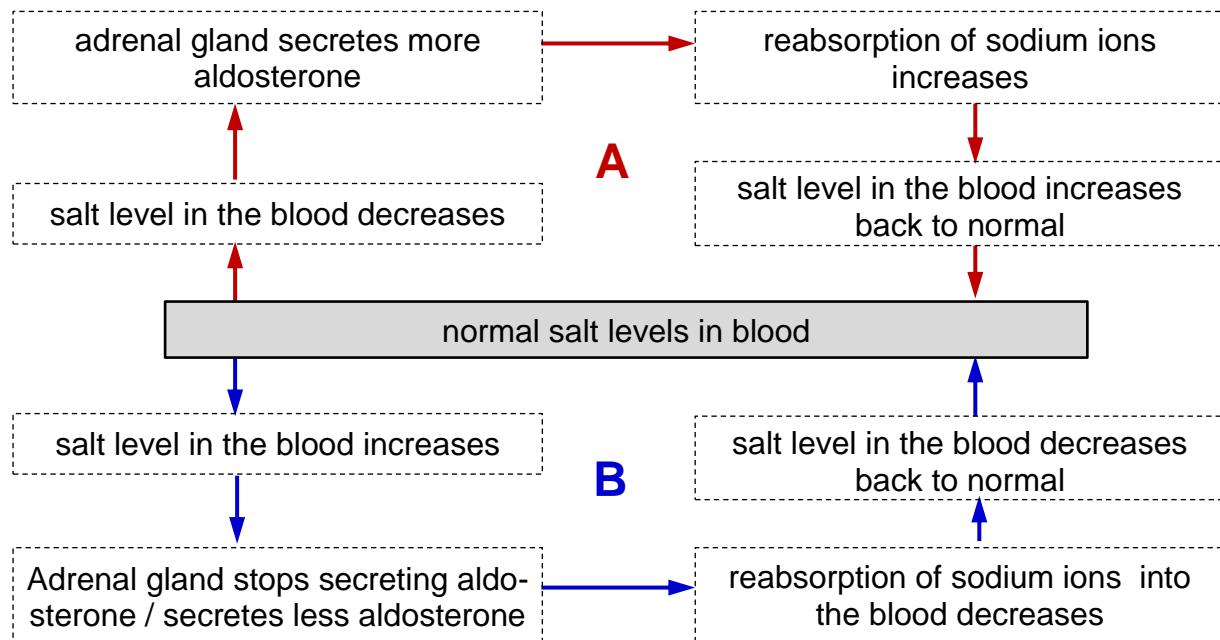


Figure 9: Negative feedback control of salt concentration in the blood

Table 7: Homeostatic control of salt concentrations

A – low salt level	B – high salt level
Low salt levels in blood and tissue fluids.	When the salt levels in the blood and tissue fluids are increased.
Receptor cells in the kidney detect decreased sodium ion levels.	Receptor cells in the kidney will detect an increased presence of sodium ions.
The adrenal gland in the kidney secretes the hormone aldosterone .	The adrenal gland stops releasing aldosterone .
Aldosterone stimulates the reabsorption of sodium ions from the filtrate and back into the blood.	Sodium ions will not be reabsorbed.
Less sodium is excreted in the urine .	More sodium is excreted in the urine .

The salt concentration of the blood and tissue fluid returns to normal and **homeostasis** is maintained.

The regulation of water and salt levels in humans:

https://www.youtube.com/watch?v=BugGCBAk_Os

Negative feedback regulating of carbon dioxide concentrations

Carbon dioxide levels in the blood affect the pH of the blood and this can have an effect on metabolic processes. CO_2 is one of the end products of cellular respiration. CO_2 dissolves in water forming carbonic acid. The more carbon dioxide there is in the blood, the more acidic the blood becomes. Changes in pH influence enzyme activity. Figure 10 and Table 8 show the bodies response to an increase in CO_2 levels in the blood.

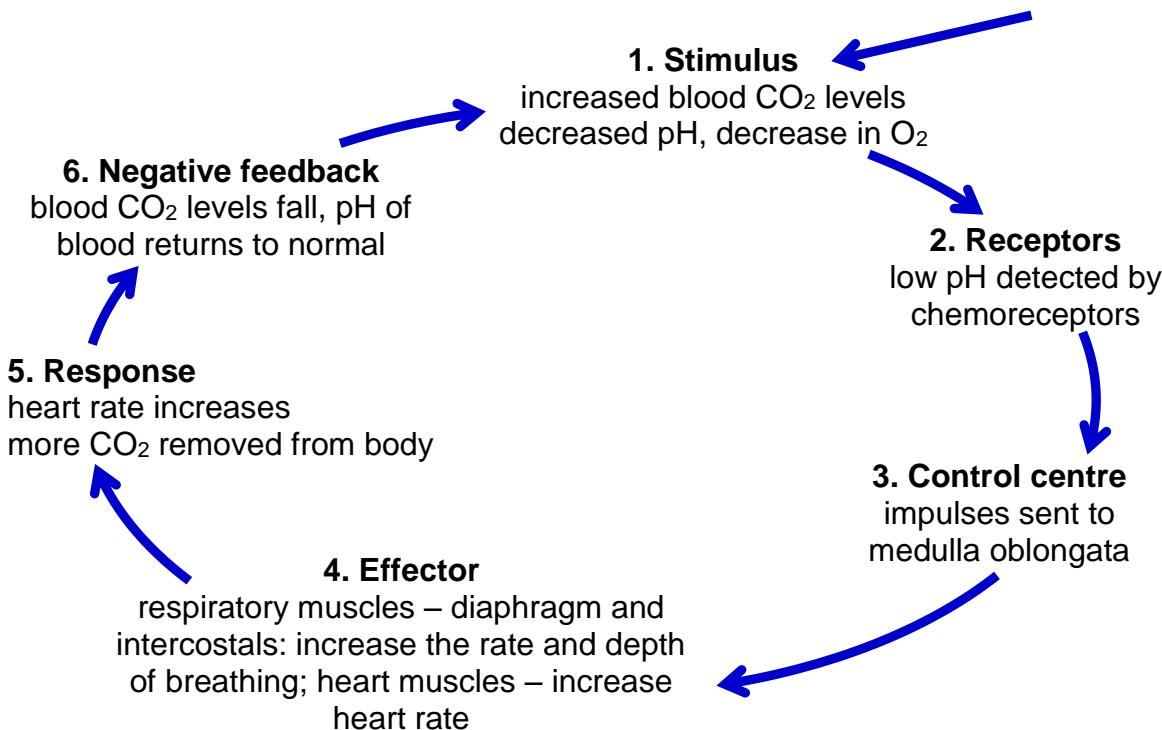


Figure 10: Negative feedback regulation of carbon dioxide levels in the blood

Table 8: Homeostatic control of blood CO_2 levels

1	High concentrations of CO_2 lead to the formation of carbonic acid. As a result, the pH of the blood will drop (the blood becomes more acidic).
2	Chemoreceptors in the carotid artery are stimulated by the drop in pH.
3	Impulses are sent to the medulla oblongata.
4	Breathing and heart muscles are targeted. Diaphragm and intercostal muscles contract increasing the rate and depth of breathing.
5	Heart rate increases.
6	More CO_2 moves to the lungs to be exhaled therefore blood CO_2 levels return to normal. Homeostasis is maintained.

Negative feedback mechanism regulating blood glucose levels

Carbohydrates are very important short-term energy storage molecules in all living organisms. In cellular respiration glucose is broken down to release energy which is stored in ATP.

The endocrine gland that secretes hormones that regulate the blood glucose levels is the pancreas. Pancreatic hormones, insulin and glucagon, work antagonistically (in opposite ways) to maintain blood glucose levels.

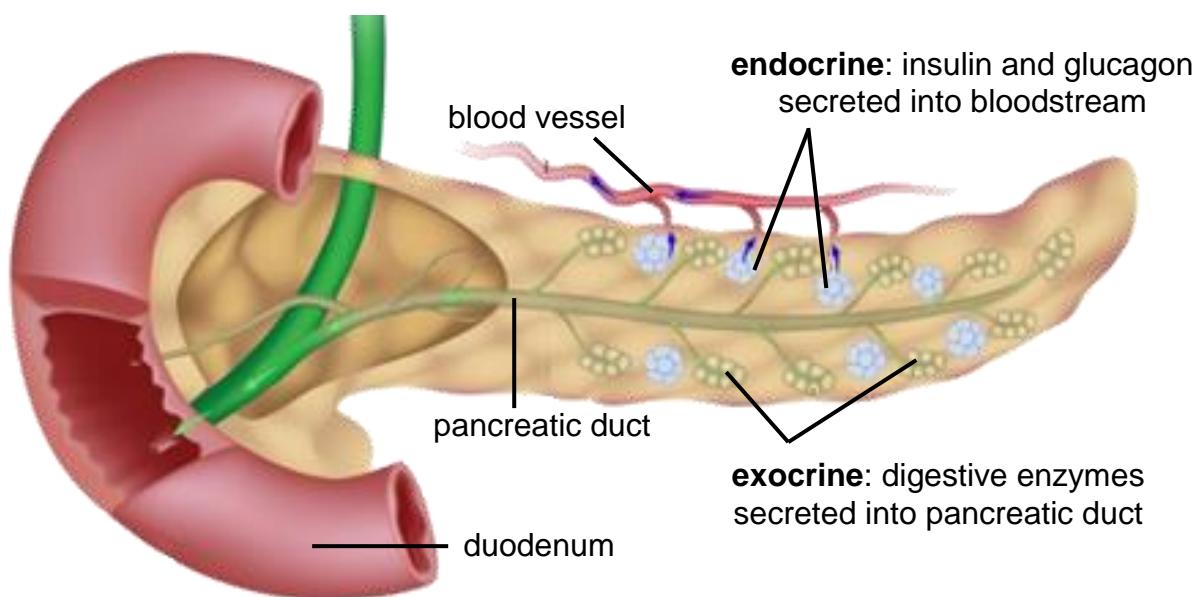


Figure 11: The pancreas showing its function as both an endocrine and exocrine gland

Blood glucose levels need to be maintained. Table 9 and Figure 12 show how:

- A **high** blood glucose levels are regulated, and
- B **low** blood glucose levels are regulated by the liver and pancreas.

Table 9: Regulation of blood glucose levels

A – high blood glucose	B – low blood glucose
Increased blood glucose levels are detected by the Islets of Langerhans in the pancreas.	Decreased blood glucose levels are detected by the Islets of Langerhans in the pancreas.
The Islets of Langerhans respond by secreting insulin into the blood-stream.	Glucagon is secreted by the Islets of Langerhans into the blood-stream.

Insulin is transported to the liver which is the effector organ.	Glucagon is transported to the effector organ, the liver .
Enzymes in the liver catalyse the conversion of excess glucose into glycogen . Glycogen is a storage carbohydrate.	Glycogen is broken down into free glucose . Glucose is released into the bloodstream.
Glucose levels in the blood return to normal .	Glucose levels are increased to normal levels.

In people with normal pancreatic functioning glucose levels are maintained and **homeostasis** is achieved.

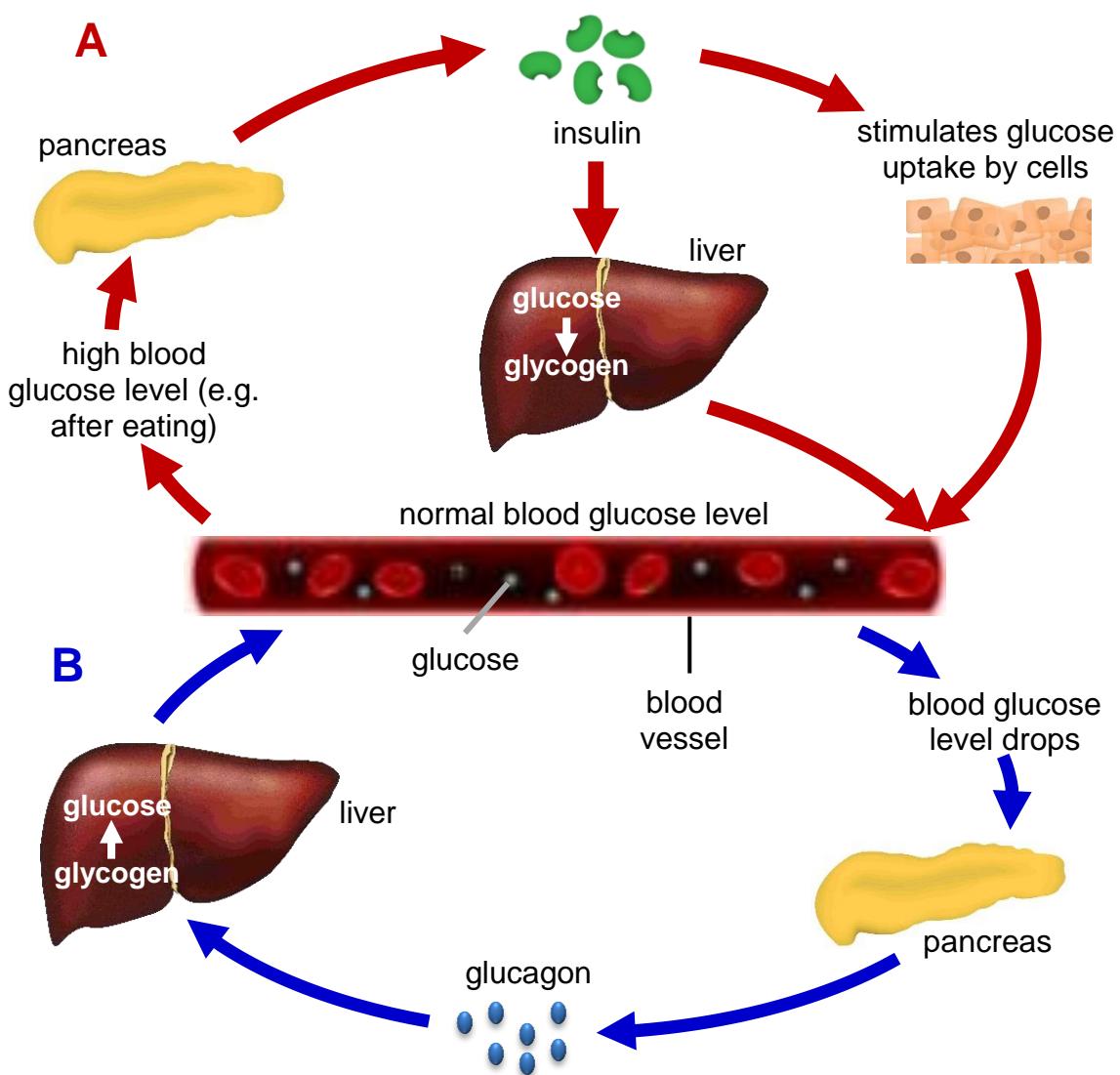


Figure 12: Regulation of blood glucose by the liver and pancreas

Negative feedback in the regulation of thyroxin levels

The basal metabolic rate (BMR) is the rate at which the body's cells use energy when at rest. Basic vital functions need to be maintained.

The hormone, **thyroxin**, produced in the thyroid gland:

- stimulates the body to increase the metabolic rate when required
- plays a vital role in the functioning of the heart and digestive system
- is important in skeletal and brain development
- is involved in maintenance of muscle tone

Iodine is an essential element needed in the production of thyroxin. A good source of iodine in our daily diet is sea salt or iodised table salt.

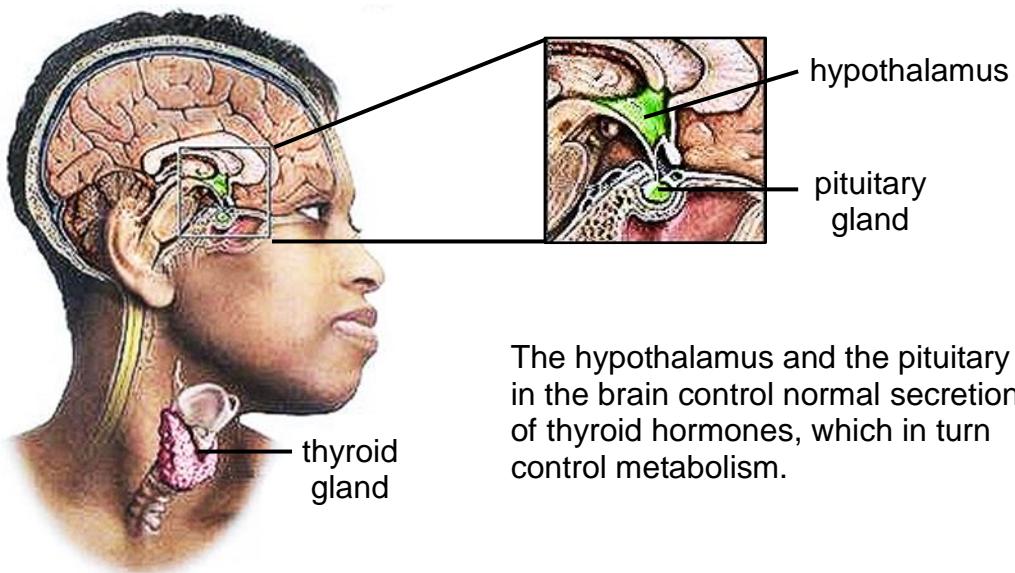


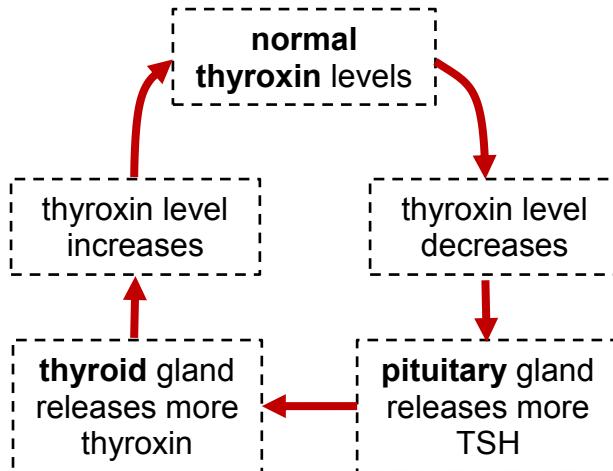
Figure 13: Location of thyroid gland in the neck
and the connection to the pituitary gland

Two glands are involved in the control of thyroxin levels:

- pituitary gland – which releases thyroid stimulating hormone (TSH)
- thyroid gland – which releases thyroxin

Homeostatic regulation of thyroxin levels is illustrated in Figure 14 and detailed in Table 10.

A – low thyroxin levels



B – high thyroxin levels

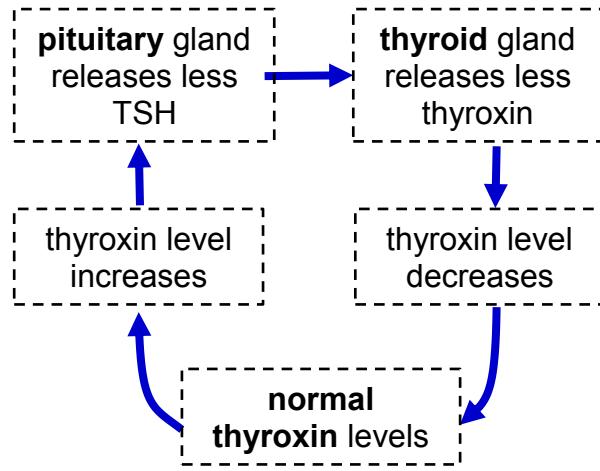


Figure 14: Flow diagram showing regulation of thyroxin levels
(A – low levels, B – high levels)

Table 10: Homeostatic control of thyroxin

A – low thyroxin level	B – high thyroxin level
When levels of thyroxin that fall below normal , this is detected by the pituitary gland .	When levels of thyroxin increase above normal, this is detected by the pituitary gland which is then inhibited.
This causes the pituitary gland to secrete more TSH .	Less TSH is secreted from the pituitary gland.
TSH is transported via the bloodstream to the thyroid gland which stimulates increased secretion of thyroxin .	Lower secretions of TSH result in the thyroid gland releasing less thyroxin .
The level of thyroxin is increased back to normal.	The level of thyroxin is decreased back to normal.

If we apply this to the negative feedback mechanism that restores thyroxin levels when it is **too low**, the appropriate steps can be made specific as follows:

Step 1: The thyroxin level in the blood decreases

Step 2: The pituitary is stimulated

Step 3: Pituitary gland increases its secretion of TSH

Step 4: TSH is transported by the blood to the thyroid gland

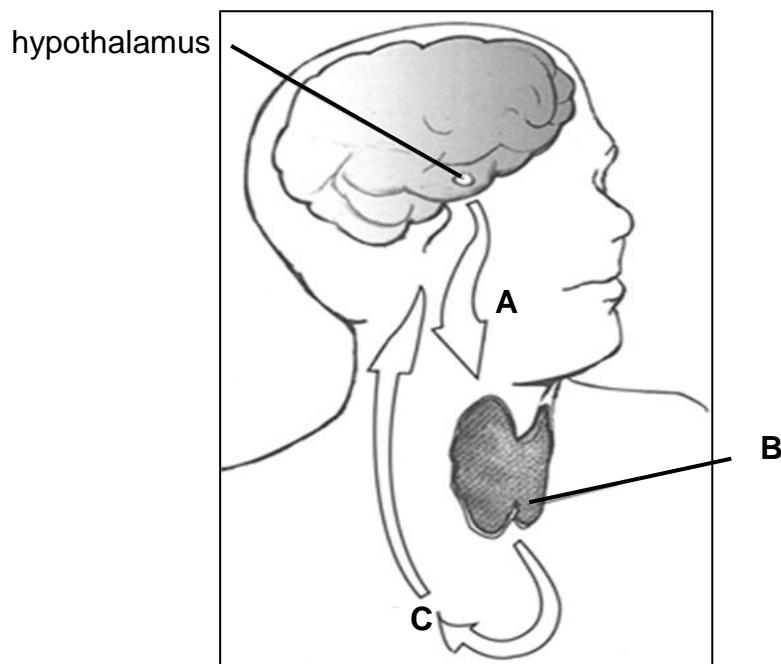
Step 5: TSH stimulates the thyroid gland to increase its secretion of thyroxin

Step 6: The thyroxin level in the blood increases

Step 7: Thyroxin levels return to normal

Activity 3: A negative feedback mechanism

The diagram below represents the interaction between two important endocrine glands. The hypothalamus is found at the base of the brain while the gland labelled B is present towards the front of the neck.



1. Provide a label for gland **B**. (1)
2. Name hormone **C**. (1)
3. State one function of hormone **A**. (1)
4. Describe the negative feedback mechanism that operates when the level of hormone **C** is higher than normal in the blood. (5)
(8)

Endocrine system disorders

In certain situations, hormone secretions are disrupted, and this affects homeostasis. Endocrine glands can either secrete too little (**hyposecretion**) or too much (**hypersecretion**) hormone. If this continues, a person would be diagnosed with an endocrine disorder.

Pituitary gland disorders

Table 11: Pituitary gland disorders

	Acromegaly	Dwarfism	Gigantism (Figure 15)
	hypersecretion of GH	hyposecretion of GH	hypersecretion of GH
Cause	too much GH secreted after puberty	too little GH produced during childhood	too much GH secreted during childhood
Symptoms	<ul style="list-style-type: none">enlargement of the hands and feetadditionally: enlargement of the forehead, jaw and nose	<ul style="list-style-type: none">well-proportioned but of short statureretarded growth and delayed puberty.	<ul style="list-style-type: none">long bones and connective tissue grow very fastperson may grow up to 2,1 to 2,5 m tallheart and other organs also enlarge, causing high blood pressure



Figure 15: Gigantism

Thyroid gland disorders

The continued production of too much thyroxin is known as hyperthyroidism (hyper = high). Continued low levels of thyroxin leads to hypothyroidism (hypo=low).

Table 12: Thyroid gland disorders

Hyperthyroidism		
Examples	Graves' disease (Figure 16A) 	Goitre (Figure 16B) 
Causes	autoimmune disease – which occurs when the immune system attacks the thyroid and causes it to overproduce the hormone thyroxin	goitre – condition linked to elevated thyroid activity
Symptoms	<ul style="list-style-type: none"> bulging eyes weight loss fast metabolism 	<ul style="list-style-type: none"> increased metabolic rate increased cardio-vascular activity increased anxiety swollen thyroid gland in neck
hypothyroidism		
Examples	Cretinism (Figure 16C) 	Myxoedema (Figure 16D) 
Cause	<ul style="list-style-type: none"> caused by lack of thyroxin from birth 	<ul style="list-style-type: none"> caused by underactive thyroid gland in adulthood
Symptom	<ul style="list-style-type: none"> physical, mental retardation 	<ul style="list-style-type: none"> mental, physical tiredness low metabolic rate increase in dermal fat roughening of skin

Pancreas disorders – e.g.: Diabetes

A person with continued (chronic) high glucose levels is said to be **hyperglycaemic**. Diabetes mellitus is a disease associated with high blood glucose levels.

The long-term effect of diabetes is damage to all the blood vessels which eventually leads to circulatory problems and multiple organ damage. Wounds on the skin do not heal well. The eyes are negatively affected, leading to poor vision and eventually blindness.

- **Type 1** diabetics might inject insulin as a treatment. There are sophisticated insulin pumps available which are directly attached to the patient and are effective in controlling glucose levels.
- **Type 2** diabetics can in most instances control their sugar levels by watching their diet, losing excess weight and exercising.

Table 13: Representation of non-diabetic and both Type 1 and Type 2 diabetics

Non-diabetic	Type 1 Diabetic	Type 2 Diabetic
Normal blood glucose levels: 80-100 mg/ml of blood	Type 1 diabetes – pancreas not producing the hormone, insulin , necessary for controlling the glucose levels in humans	Type 2 diabetes – insulin is available, but the body is unable to control the glucose levels

Table 13 and Figures 17A to 17B illustrate the normal insulin glucose interaction, and what happens in a Type 1 or a Type 2 diabetic.

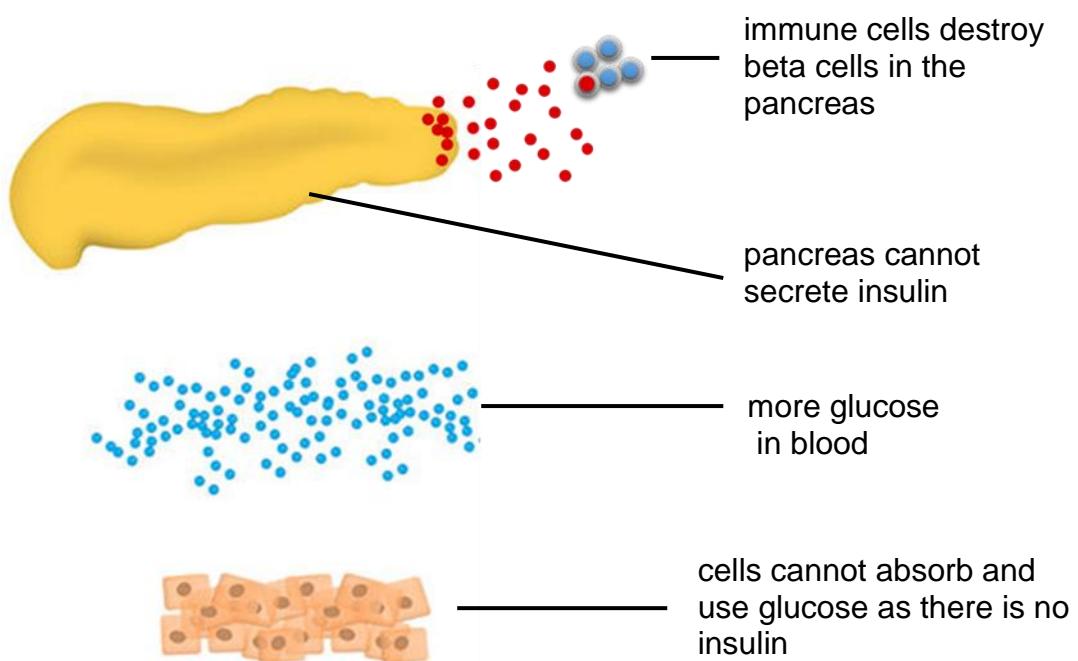


Figure 17A: Type 1 diabetic – no insulin

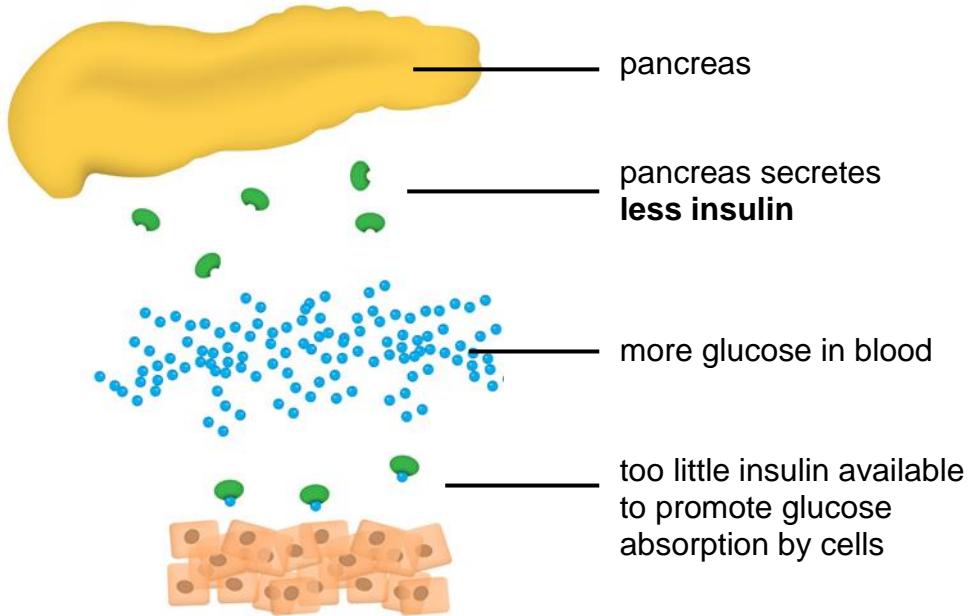


Figure 17B: Type 2 diabetic – less insulin produced

Activity 4: Research task on endocrine disorders

You will need to research an endocrine disorder caused by the hypersecretion or hyposecretion of an endocrine hormone. You will present your information in either a PowerPoint Presentation or a poster format. The following needs to be covered:

1. What hormone is involved with this disorder?
2. Where is the hormone produced?
3. What are the target organs/structures of the hormone?
4. How is the secretion of the hormone regulated/controlled?
5. What is the normal function of the hormone?
6. How does the hormone contribute to homeostasis?
7. What are the causes of hypersecretion or hyposecretion?
8. What are the symptoms and effects of hypersecretion or hyposecretion?
9. What are the treatments for under or over activation of the hormone pathway?

Your teacher will provide you with a mark scheme. N.B.: Citation of at least 3 references.