

YEAR 12 ATAR COURSE REVISED EDITION



**ACADEMIC
TASK FORCE**

REVISION SERIES

HUMAN BIOLOGY

~~~~~ UNITS 3 & 4 ~~~~~



**ANT MECZES**



**ACADEMIC  
TASK FORCE**

REVISION SERIES

# **HUMAN BIOLOGY**

YEAR 12 ATAR COURSE  
UNITS 3 & 4

SECOND EDITION

**ANT MECZES**



**ACADEMIC GROUP**

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#### **About the Author**

Ant began his teaching career in 1994. He taught in Newcastle-upon-Tyne, UK, before moving to Australia in 2007. He is a passionate Science educator who has spoken at a number of conferences. Ant is Head of Science of Perth Modern School and has been at the school since 2009.

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This book is dedicated to my wife, Emma, and my children, Holly and Luke, who gave me the time and space to write it. Also to my Human Biology Y12 students, especially Lydia, Caitlyn, Trinity and Katherine, who gave me helpful feedback on some sections. You were the inspiration and motivation to complete the work!

Ant Meczes, July 2017

# UNIT 3 AND 4 REVISION GUIDE

## INTRODUCTION

The first thing to recognise is that the human body is a coordinated series of systems. Consequently, an idea in one chapter may be present in another chapter, even though it is not obvious. Chapters are written to bring order to a body of knowledge. However, if we want to truly achieve success, we need to see the unifying themes flowing through the chapters. This deeper, conceptual understanding enables us to have a greater grasp of the knowledge presented. It allows us to move from the surface understanding of knowledge and comprehension to the deeper understanding of abstract ideas, leading to application of understanding and synthesis.

The purpose of this revision guide is to identify the key ideas of each chapter and to introduce you to some of the underlying themes which will hopefully help you to improve and refine your revision techniques in preparation for the final examination.

## FIRST PRINCIPLES

### 1. Understand the structure of the paper.

The paper is written in three parts; multiple choice (30 marks), 6–10 structured questions (100–110 marks) and extended questions (40 marks); 2 from Unit 3 and 2 from Unit 4, of which you choose **one** from each section.

The weighting for each section is as follows: Multiple choice (30%), structured questions (50%) and extended questions (20%)

You are given 10 minutes reading time at the beginning of the exam. During this time, you are not allowed to write anything. Most people begin at the start of the paper and read through. This is not the best approach. You have only one section where there is choice; the extended section. Here you are expected to complete **TWO** out of three questions. Each question is worth 20 marks and is usually divided in two to four parts. The key is to read **ALL** the questions and its parts during the reading time and determine how many marks you can get roughly for each question. This will help you decide which the two best questions for you to attempt are. Remember, you may not be able to answer part a). However, you can answer b) and c) and these are worth more marks. The common mistake is to start an extended question and realise that you can only answer the first section. Disaster strikes and panic sets in. Avoid this by reading the extended section first.

### 2. Read a chapter on a set topic and then attempt questions on that topic.

Many students read the chapter of textbooks and make the mistake of simply colouring it in. This doesn't mean that you have read it. One of the most effective ways of making notes is to MindMap®. There are videos of this on YouTube. Whilst both MindMapping and note-taking are effective methods of revision the night before a test or exam, MindMapping shows significant improvements when a test on the same topic taken 6 weeks later. This is because it is an active way of making notes. Sometimes, a routine such as Generate – Select – Connect – Elaborate can make this a more effective process. Other effective note-taking strategies include:

- See – Think – Wonder routine: I see this in the text. I think this about the text. I wonder this about the text.
- Think – Puzzle – Explore: I think this as I read the text. I am puzzled about this. I would like to explore this further (if I have time).

These strategies are aimed at active engagement with texts or videos.

### 3. The three keys to exam success

Simply put, these are as follows:

- **Knowledge:** Know your stuff. Read the material and read it again until you understand it.
- **Practice of exam questions:** The more you practice, the more familiar you will become with different styles of questions and how questions are written to access the knowledge you know. The key to answering exam questions is to identify what they are asking and demonstrate understanding, both of the questions and the knowledge learnt, including application of this knowledge.
- **Confidence:** This comes through the first two. You need to believe you can succeed. You can succeed if you know your stuff and you have tested yourself on it, by practising exam questions.

## CHECKLISTS

At the beginning of each chapter is a checklist of the course objectives. Each objective is written in the first person so that you take ownership of them. There are three columns following the checklist. The first column is your first read through. The second is to ensure you have revised the material and practised questions on the material. The final column is to build confidence in you; if you can teach someone else the objectives, it shows you truly understand the material and can articulate it to others. Also, it ensures that you have covered the material **AT LEAST** three times.

## EXTRA INFORMATION / READING

The information contained within the grey boxes is extra information / reading. It is not in the syllabus and therefore not examinable. However, it does provide context and allows students who want a deeper understanding to go a little further.

Sometimes grey boxes contain exam tips. These are clearly labelled.

Be aware that there have been some minor changes to the syllabus. The syllabus is your starting point, not the textbook. Sometimes there is additional information that goes beyond the text or things are put differently. Use a range of resources to develop your understanding. Then, discuss what you have learned with others, including your peers.

# SCIENTIFIC INQUIRY (UNIT 3)

# 1



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Done | Revise | Teach others |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Chapter 1: Science Inquiry Skills (SIS) (Unit 3 and 4)</b>                                                                                                                                                                                                                                                                                                                                                                                                            |      |        |              |
| <ul style="list-style-type: none"> <li>I can identify, research and construct questions for investigation; propose hypotheses; and predict possible outcomes</li> </ul>                                                                                                                                                                                                                                                                                                  |      |        |              |
| <ul style="list-style-type: none"> <li>I can design investigations, including the procedure(s) to be followed, the materials required, and the type and amount of primary and/or secondary data to be collected; conduct risk assessments; and consider research ethics</li> </ul>                                                                                                                                                                                       |      |        |              |
| <ul style="list-style-type: none"> <li>I can conduct investigations safely, competently and methodically for the collection of valid and reliable data</li> </ul>                                                                                                                                                                                                                                                                                                        |      |        |              |
| <ul style="list-style-type: none"> <li>I can represent data in meaningful and useful ways, including the use of mean, median, range and probability; organise and analyse data to identify trends, patterns and relationships; discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and the sample size may influence limitations in data; and select, synthesise and use evidence to make and justify conclusions</li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can interpret a range of scientific and media texts, and evaluate models, processes, claims and conclusions by considering the quality of available evidence, including interpreting confidence intervals in secondary data; and use reasoning to construct scientific arguments</li> </ul>                                                                                                                                     |      |        |              |
| <ul style="list-style-type: none"> <li>I can select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions.</li> </ul>                                                                                                                                                                                                                              |      |        |              |
| <ul style="list-style-type: none"> <li>I can communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports.</li> </ul>                                                                                                                                                                                                                                                          |      |        |              |

In the exam, there is usually a question in the structured section worth between 8 and 14 marks.

**Helpful hints:**

- The question will often contain an investigation which is unfamiliar to you, but an area of actual research.
- Pay careful attention to the introductory blurb. Identify the key points.
  - What factors demonstrate that it is a controlled investigation?
  - What is the 'factor', i.e. variable being investigated?
  - Is there any use of data? Take note of this and be prepared to refer to data in your answer.
- Scientific inquiry is a fundamental skill within Science and follows the same principles from K-12 and beyond. Therefore, Unit 3 and 4 build on the Science Inquiry skills learnt in Units 1 and 2.

**Definition of Terms and Key Points:**

- **Independent variable:** The one that is changed by the experimenter, whilst the other variables are kept the same.
- **Dependent variable:** The one that is measured and changes (or not) as a result of the independent variable. Note, that a percentage change has been analysed and so isn't the dependent variable, but some tables have this information on them.
- **Control variables:** A variable is a factor that has the potential to be changed. Changing more than one factor in an investigation will make the data invalid, because it is not certain which factor caused the effect (or non-effect) if more than one is changed. Therefore, control variables are factors that can change, but are kept the same to ensure that the experiment is a **fair test**, i.e. these factors are kept the same in both the control and the experimental groups in an investigation.
- **Uncontrolled variables:** Some factors are not possible to keep the same for either the experimental or control group. In these situations their impact needs to be minimised as far as is possible.
- **Control experiment:** The best experiments have two or more set ups whereby the only difference is as a result of changing the variable being tested. This allows comparisons to be made, so that any difference in the results is down to the variable being tested/ investigated.
  - Placebos, i.e. tablets or substances that have all the same properties as the actual tablet or substance, except the active ingredient, can be used to show the differences between the actual variable being tested (if there are any) and psychosomatic effects. Often we find that changes can occur due to psychosomatic effects (mind having an affect over the body), but it is the **difference** between the control and experimental group that allows the experimenter to see whether there is a significant difference or not. Ideally, a statistical test would be conducted to identify the degree of significance or not.
  - **Blind and double blind.** A blind experiment is aimed at preventing bias. The subjects in an investigation should not know if they are receiving the test substance or a placebo. The best experiments are double blind, whereby an independent person organises the administration of the substance to the subjects. This way it avoids unintentional bias from the experimenter who is also unaware of who is receiving the placebo or the active substance. This results in more reliable data.
- **Hypothesis:** A testable prediction. It is usually written showing the relationship between the independent and dependent variable. E.g. As the concentration of the enzyme is increased (independent variable), the time taken for solution to go clear will decrease (dependent variable). It may be written as an "If/Then" statement.
  - The best investigations are **quantitative**, i.e. they involve numerical measurements, rather than **qualitative**, i.e. observations that don't involve numbers or measurements, because statistical analysis can be conducted.

- The following terms are often muddled:
  - **Precision:** The degree of detail allowed by the actual experiment. If data is recorded to two decimal places, then the mean can only be recorded to a maximum of two decimal places. It can't be greater than this, because there isn't this degree of precision in the measurements recorded by the apparatus used.
  - **Reliability:** A measure of the extent the experiment gives the same result every time it is repeated. Experiments should be repeated at least 3 times, preferably more. This allows **outliers** or **anomalous results** to be identified or removed. If the investigation is repeated only twice and there is a difference in the two results, then it is not possible to know which result to use. Experiment increases in reliability with greater repetition. Reliability results in consistency of data.
    - **Repetition:** doing the experiment many times using the same equipment (and/or subjects), is the main way that reliability is achieved.
    - **Replication/Duplication:** This can be using a large number of subjects involved in the investigation, i.e. the sample size is large to account for **biological variance** and allow differences between subjects to be reduced (experimental error) in their effect on the overall conclusions. Also, replicates can be having a number of identical experiments running at the same time.
  - **Accuracy:** This is the closeness to the true value. In an investigation it is possible to have reliable results, but they are not accurate. For example, if I threw darts at a dart board and consistently hit double three, my result is reliable. But, if the true value is the bullseye (the centre of the board), my throwing lacks accuracy. Most of the time our results are reliable, but as we may not know the true value, they are not accurate. These two terms are most commonly muddled.
    - **Secondary data:** This is useful for us to determine the accuracy of the investigation collected. In order for most investigations to be verified another person needs to have followed the same procedure and come up with similar results. As it has been collected independently it serves to provide supportive evidence for the experiment currently being conducted.
    - **Literature reviews:** It is good practice when conducting an investigation to see whether it has been done before. Conducting a review of the subject under consideration is an excellent way to develop procedures and find secondary data, which can either support or offer an alternative to the data collected in the current investigation.
  - **Validity:** The investigation tests the question being asked. It can only do this if it has been set up fairly, with precision and reliably, otherwise, it is not valid.
- **Risk assessments:** Whenever an investigation is conducted, safety needs to be considered. The experimenter needs to identify the potential dangers to the participants/subjects involved. Where possible, the aim is to reduce the risks to a minimal.
- **Ethics:** These are moral principles or practices that govern the conduct of the investigation. **Ethical behaviour** follows these principles and procedures. In the case of investigations, participants need to be made fully aware of the procedures and possible side effects or risk of harm prior to their participation, which is voluntary. This results in informed consent. The identity of the participants is kept confidential, i.e. their details are not revealed except to the people directly involved in the study. This is not the same as privacy, although these terms are often confused.
- **Scientific methodology:** This comes out of a systematic approach to answering questions that arise from observations made about the physical and natural world.
  - An observation is made which leads to a question posed or problem to solve.
  - As much information as possible is collected with regards to the question posed.
  - A hypothesis is proposed which presents a possible solution to the question based on the information presented. This hypothesis needs to be testable.
  - An experiment is set up to test the hypothesis.

- o Data is collected, presented and analysed.
- o This results in conclusions, which either support or disprove the hypothesis. (In Science, unlike in Maths, no hypothesis can be proved. The data can provide evidence to support the hypothesis. At a later date, more data may become available which disproves the current thinking and offers an alternative theory).

### Tables and Graphing

- Initially, data is best presented in a table. Then, in order to see the patterns and trends, a graph is useful. **Trends** are overall observations that the data set shows, whereas **patterns** are repeatable observations in the data.
- Tables should include the following:
  - o A title. So often students forget this. What is the table about? State it. Where possible, link the independent to the dependent variable in the title.
  - o Columns. The first column should have the independent variable in it, with the columns that follow containing the dependent variable. Each column should be **labelled** with the **units** at the top of the columns, not imbedded in the table.
  - o Totals and means. Whilst many tables simply have the means without the totals, it is good to show where the means are derived from. Therefore, I recommend including both, especially if outliers have been removed from the totals.
- Graphs are usually worth between 4-6 marks. They should include the following:
  - o A title. What does the graph show? State the relationship between the independent and dependent variable.
  - o Axes.
    - The independent variable is **always** on the x (horizontal) axis.
    - The dependent variable is **always** on the y (vertical) axis.
    - Ensure you keep the same scale throughout the graph, i.e. equal intervals of units.
    - Label each axis and **include units** where possible.
  - o Scale. The graph should cover more than half of the paper provided; otherwise it is a poor scale.
  - o Line graphs. Plot the points and join each point using a ruler. This differs from graphs in Chemistry and Physics where the line/curve of best fit (trend line) would be drawn.
  - o Key. When comparing two categorical data sets, e.g. control v experimental group, a key is used to distinguish between the two lines drawn. Usually different colours would be used for the lines
  - o Reading from a graph: The value you read is a precise point on the graph, not an approximate value, so don't use ~ before your answer.





## QUESTIONS

20 marks

A student was investigating speed of reaction. He wanted to see if his left hand was faster than his right hand at catching a ruler. He was left handed. He was to catch the metre ruler between his thumb and first finger, which one of his friends measured to ensure they were always 2 cm apart before the ruler was dropped. His friend always dropped the ruler from the same point; level with his fingers. The ruler wasn't touching either of his fingers before it was dropped. The student was sat down. He used a conversion table to work out the reaction times and recorded the results in the table below:

A table to compare reaction times between the left and right hand of left handed individual

|        | Reaction times (s) |         |         |         |         |       |         |
|--------|--------------------|---------|---------|---------|---------|-------|---------|
| Handed | Trial 1            | Trial 2 | Trial 3 | Trial 4 | Trial 5 | Total | Average |
| Left   | 0.10               | 0.15    | 0.08    | 0.11    | 0.17    |       |         |
| Right  | 0.35               | 0.40    | 0.29    | 0.34    | 0.10    |       |         |

1. Complete the totals and averages, but remove any outliers from the data. [3]

2. What would have been a suitable hypothesis for this investigation? [1]

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3. What was the independent variable? [1]

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4. What was the dependent variable? [1]

---

5. Name two variables mentioned that the student controlled. [2]

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6. How did the student make the investigation reliable? [1]

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7. Write a conclusion for this investigation based on the hypothesis? [3]

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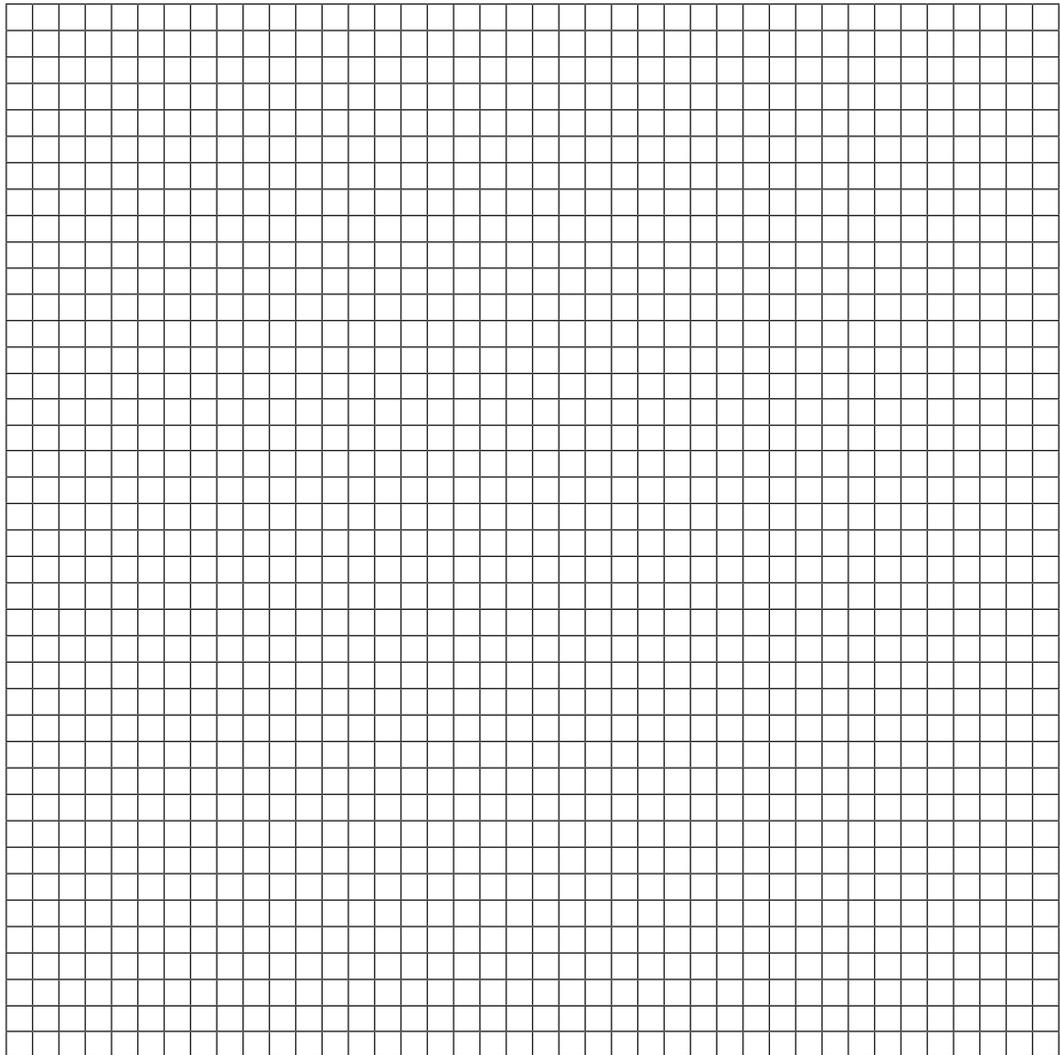
8. A friend of the students conducted a similar experiment. However, he used 15 right-handed and 15 left-handed students. Explain why you think this would result in a more reliable conclusion? [2]

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9. Draw a graph of the results. [6]



# HOMEOSTASIS AND THE NERVOUS SYSTEM (UNIT 3)

2



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Done | Revise | Teach others |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Detecting and regulating change (Introduction to homeostatic mechanisms) (Unit 3)</b>                                                                                                                                                                                                                                                                                                                                                                                      |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain that homeostatic processes involve nerves and hormones in maintaining the body's internal environment within tolerance limits through the control of metabolism and physiological and behavioural activities.</li> </ul>                                                                                                                                                                                                 |      |        |              |
| <ul style="list-style-type: none"> <li>I can describe that the reflex arc comprises of specially structured neurons, including sensory, interneuron and motor neurons, to transmit information from the receptor to the effector to respond rapidly to stimuli.</li> </ul>                                                                                                                                                                                                    |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how transmission of nerve impulses is via electro-chemical changes that occur at the generation of the impulse, the propagation of the impulse along the nerve fibre, and the transfer of the impulse across the synapse.</li> </ul>                                                                                                                                                                                     |      |        |              |
| <ul style="list-style-type: none"> <li>I can name the different receptors that detect changes in the internal and external environments, including thermoreceptors and osmoreceptors.</li> </ul>                                                                                                                                                                                                                                                                              |      |        |              |
| <b>Central and Peripheral Nervous System (Unit 3)</b>                                                                                                                                                                                                                                                                                                                                                                                                                         |      |        |              |
| <ul style="list-style-type: none"> <li>I know that the structure and function of the divisions of the nervous system can be observed and compared at different levels in detecting and responding to the changes in the internal and external environments including:                             <ul style="list-style-type: none"> <li>central-peripheral</li> <li>afferent-efferent</li> <li>autonomic-somatic</li> <li>sympathetic-parasympathetic</li> </ul> </li> </ul> |      |        |              |
|                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |      |        |              |

| Key teaching points                                                                                                                                                                                                                                                                                                                                                      | Done | Revise | Teach others |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <ul style="list-style-type: none"> <li>I understand that the nervous and endocrine systems work together to co-ordinate functions of all body systems, but differ in terms of: <ul style="list-style-type: none"> <li>speed of action</li> <li>duration of action</li> <li>nature and transmission of the message</li> <li>specificity of message</li> </ul> </li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can name the parts of the central nervous system, including the brain (cerebrum, cerebellum, medulla oblongata, hypothalamus, corpus callosum) and spinal cord, and explain the specific roles of each in the co-ordination of body functions</li> </ul>                                                                        |      |        |              |
| <ul style="list-style-type: none"> <li>I can describe how the CNS is protected by bones of the skull, the meninges and cerebro-spinal fluid.</li> </ul>                                                                                                                                                                                                                  |      |        |              |

### Helpful hints:

- The nervous system follows the same pattern of response as the endocrine system:
- The hardest concept to understand is propagation along a neuron, i.e. how action potentials occur along neurons. Therefore, it is worth watching short videos and animations on this on the Internet.
- Try and write in an ordered, structured fashion, using precise language. For example, the saltatory conduction is movement along the neuron from node to node.
- Use of precise language is important. In both unmyelinated neurons and myelinated neurons the impulse travels along the entire length of the neuron. The difference is that in myelinated neurons, the impulse “jumps” from node to node, whereas in unmyelinated neurons the impulse conducts along every part of the membrane, which explains why it is slower. (More detail below)
- Understand the difference between describing a structural neuron and a functional neuron.

### Overview:

- Homeostasis comes from the words *Homeo-*, which means “the same”, and *-stasis*, which means a state or condition that remains unchanged. The actual definition used is: Maintaining a constant or stable internal environment, despite changes to the internal or external environment.
- Changes to the environment are called **stimuli** and these are detected by **receptor cells**.
- The Central Nervous System (CNS): Composed of the brain and spinal cord.
- The Peripheral Nervous System (PNS): Composed of:
  - The **afferent (sensory)** division
    - Somatic sensory neurons: **from** skin and muscles.
    - Visceral sensory neurons: **from** internal organs, e.g. heart, lungs.
  - The **efferent (motor)** division
    - Somatic motor division: **to** skeletal muscles.
    - Autonomic division: **sympathetic**: dominant in fight or flight scenarios.  
**parasympathetic**: results in return to normal functions.
- Summary of pathway of stimulus to response:  
Stimulus → receptor → (sensory neuron) → modulator → (motor neuron) → effector → response.
  - Receptor cells are found in the **sense** organs.
  - The stimulus is **transduced** into electrical impulse which **disrupts the** membrane of sensory neuron → **action potential** *if* stimulus is strong enough to cross the *threshold level*.
  - Modulator (Coordinator) is the CNS. Spinal cord → reflex. Brain → “Decides what to do”

- o Effector = muscle or gland. E.g. muscle contracts, gland secretes fluid.
- Many responses involve both **hormonal** and **nervous** control.

## NEURONS

- Neurons are single nerve cells. As cells, they contain all the organelles found in other types of cell, including a cell surface membrane, cytoplasm and a nucleus. As well as the nucleus, the other organelles are found in the **cell body**. (Sometimes the cell body is called the soma.)
- As with many cells, nerves are adapted to their function. They are fundamentally specialised cells. Their structure is related to their function:
  - o **Cell body (Soma)**: The part of the neuron that contains the nucleus.
  - o **Dendrites**: extensions on the cell body of the neuron. Allow communication with other neurons. Transmit/conduct impulses towards the cell body.
  - o **Axons**: elongated sections of cytoplasm. Transmit messages over long distances, away from the cell body.
  - o **Myelin sheath**: lipid (fatty tissue) produced by Schwann cells, which surround the axon on some neurons (**myelinated neurons**). Gaps in the myelin sheath speed up conduction by allowing the impulse to “jump” from one node to the next. This is called **saltatory conduction**. The gaps are known as **nodes of Ranvier**.
- Neurons form nerve fibres (a term which usually refers to the cytoplasm). Nerve fibres are bundled together into nerves (nervous tissue – held together by connective tissue).

### Types of Neurons:

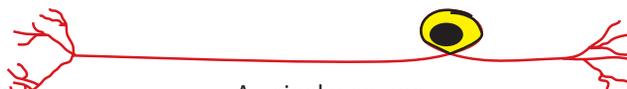
There are different types of neuron:

#### 1. Functional:

- **Sensory (afferent)** – carry impulses **towards** CNS.
- **Motor (efferent)** – carry impulses **away** from CNS.
- **Inter/Connector/Association/Relay** – link sensory to motor within grey matter of spinal cord.

#### 2. Structural:

- **Unipolar** – have just one extension from cell body. Cell body on one side of axon. All sensory neurons.
- **Bipolar** – have one axon and one dendrite. Occur in the eye, ear and nose.
- **Multipolar** – have one axon and multiple dendrites. Most common. They include interneurons in brain and spinal cord, and motor neurons to the skeletal muscles.

| Structural Type of Neuron | Diagram                                                                                                                                               | Examples                       |
|---------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------|
| Multipolar                |  <p>A multipolar neuron<br/>E.g. motor/efferent neuron</p>        | motor/efferent neuron          |
| Bipolar                   |  <p>A bipolar neuron<br/>E.g. in the eyes</p>                     | in eyes                        |
| Unipolar                  |  <p>A unipolar neuron<br/>E.g. most sensory/afferent neurones</p> | most sensory/afferent neurones |

## Transmission of a nerve impulse

- The nervous system performs three functions:
  1. To collect information about the internal and external environment.
  2. To process and integrate information, often in relation to previous experience.
  3. To act on the information received, sometimes involuntarily, as in a reflex, or as a result of a decision.
- The functions described above are conducted at a remarkable speed resulting in fast responses to stimuli.
- The transmission of a nerve impulse along a neurone is one of the hardest concepts to understand. There are three key ideas that need to be understood; resting potential, action potential and refractory period.

### a. Resting potential

- When a neuron has not been stimulated it is said to be at rest. It is in its normal state.
- The inside of the cell membrane is negative compared to the outside, which is positive. Unlike charges are attracted to each other. If these charges were to come together, they would generate energy. As the charges are separated by a membrane, the energy is not released. It is **potential energy**. This difference in energy on either side of the membrane can be measured. At rest, it is  $-70\text{mV}$ . Therefore, this is known as the **resting potential**.

#### *Extra information*

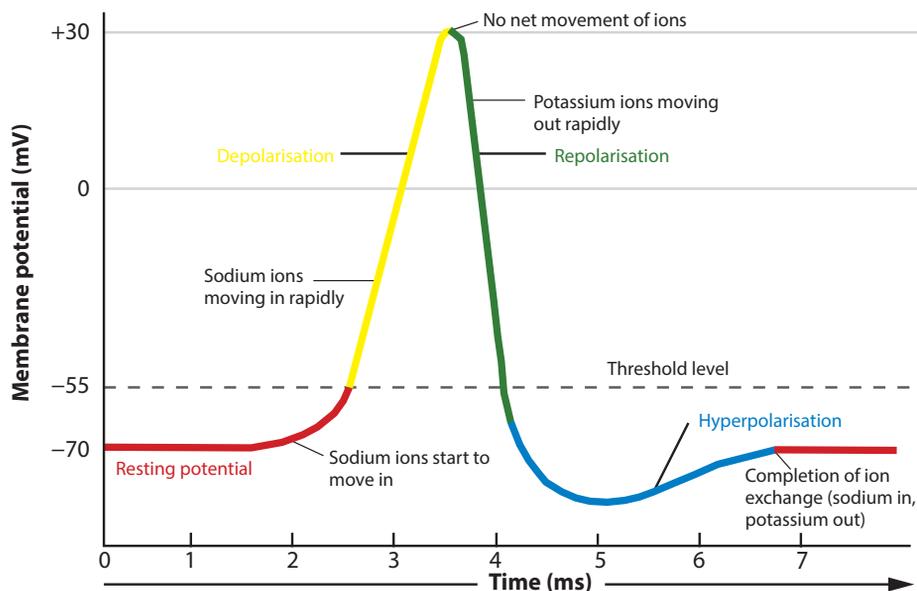
- The resting potential is mainly controlled by the distribution of four ions: potassium ( $\text{K}^+$ ), sodium ( $\text{Na}^+$ ), chloride ( $\text{Cl}^-$ ) and organic anions ( $\text{COO}^-$ ).
- At rest, there are more sodium and chloride ions on the outside of the membrane, and more potassium and organic anions on the inside.
- The membrane is more permeable to potassium ions, which leave the neuron down their concentration gradient, by diffusion. Initially the potassium ions are about 30 times greater on the inside than on the outside of the membrane, so this results in the membrane becoming more negative on the inside.
- As the number of potassium ions increases on the outside, it becomes more difficult for them to leave.
- A point is reached whereby the number of potassium ions moving into the cell equals the number of potassium ions moving out, i.e. equilibrium has been reached.
- Therefore, it is the potassium ions that are largely responsible for the creation of a resting potential.

- The difference in concentration of ions across the membrane is maintained by the active transport of ions across the membrane against the concentration gradients. The mechanism for conducting this process is carried out by **pumps**. The main pumps are **sodium-potassium pumps**. These pumps move three sodium ions out of the cell, for every two potassium ions moved into the cell, using energy from ATP.

### b. Action potential

- At rest, the membrane is said to be **polarised**.
- When the membrane receives an appropriate stimulation, the negative charge inside the membrane at a specific point becomes positively charged. This change in charge is as a result of an **action potential** and the membrane is said to be **depolarised**.
- In order for the neuron to depolarise, it needs to be stimulated enough for it to pass the **threshold level**. This is at about  $-55\text{ mV}$  and is caused by the influx of some sodium ions, due to increased permeability (A few sodium-ion gates/channels have opened).

- Once this point is reached, the action potential remains constant, regardless of the size of the stimulus, i.e. the action potential is either generated or not. It is an **all or nothing** response.
- The size of the action potential doesn't decrease as it is transmitted along the membrane, but always stays the same.
- Once the threshold has been passed, sodium ion gates open rapidly, increasing permeability to sodium ions. This allows the sudden influx of sodium ions into the cell at this point, by diffusion. This causes the membrane to become more positively charged, i.e. depolarised.
- The potassium ion gates take longer to open, but eventually do, just as the second part of the sodium ion gate begins to shut. At this point potassium ions begin to leave the cell.
- At about +30 mV, the sodium ions entering are equal to the potassium ions leaving, so there is no net movement of ions.
- With the sodium ion gates shut and the potassium ion gates open, the membrane begins to become more negatively charged again. It is being **repolarised**.
- The potassium ion gates eventually shut, but it does overshoot slightly causing **hyperpolarisation** before returning to the resting state.



### c. Refractory period

- After an action potential, the potassium ions quickly restore the resting potential.
- However, the sodium ions diffusion into the cell is prevented in that region of the membrane.
- So, during the action potential and for about 1 millisecond afterwards, further action potentials cannot be generated in that part of the membrane. This is known as the **refractory period**.
- Consequently, action potentials can only be propagated/transmitted in the region that is not in refractory, which causes the impulse to travel in one direction only.
- When the refractory period has passed, a further action potential can be carried out in the same nerve, because the first impulse has travelled further down the neuron.
- This means the refractory period effectively sets the limit of frequency of impulses along a neuron.

**Extra information**

- There are two types of refractory period; **absolute** and **relative**.
  1. **Absolute refractory period** lasts for about 1 ms during which no new nerve impulses can be propagated, regardless of how intense the stimulus.
  2. **Relative refractory period** lasts for about 5 ms during which new impulses can only be propagated if the stimulus is more intense than the normal threshold.

**d. Transmission of the impulse along the entire length of the neuron**

- Two main factors influence the speed of conduction along a neuron:
  1. **The diameter of the axon:** The greater the diameter, the faster the speed of transmission.
  2. **The myelin sheath:** Produced by **Schwann cells** this fatty material (lipid) is unable to conduct electricity, so an action potential can only form at the gaps in the myelin sheath. These gaps or **nodes of Ranvier** are about 1-3 mm apart. Action potentials are set up at these nodes and, effectively, conduction 'jumps' from node to node. This is called **saltatory conduction**.
- In non-myelinated neurons, an action potential is generated in the next section of the membrane due to the fact that there is a difference in current between the area where the action potential is occurring and the neighbouring section of membrane.

**Note:** It is not a flow of electrons like an electric current, but rather the signals are very brief changes in the distribution of electrical charge across the plasma membrane, caused by the very rapid movement of sodium and potassium ions into and out of the axon).

- The size of a nerve impulse is the same regardless of the size of the stimulus. Differences in intensity are generated in two ways:
  - o The number of nerve fibres depolarised: A strong stimulus causes more of the nerve fibres to be depolarised compared to a weak stimulus (providing the weak stimulus is strong enough to pass the threshold level).
  - o The frequency of nerve impulses: A strong stimulus results in more nerve impulses in a given time than a weak stimulus (providing the weak stimulus is strong enough to pass the threshold level).

**Exam tip:** When asked to differentiate between an unmyelinated and myelinated neuron, many students will state that the nerve impulse in an unmyelinated neuron travels down the whole length of the neuron. Of course, this is also true of a nerve impulse in a myelinated neuron. What they mean is that in an unmyelinated neuron conduction occurs across every adjacent section of the membrane along its entire length, whereas in a myelinated neuron, saltatory conduction occurs, so sections of the neuron are missed out.

**e. Transmission between neurons (Synapses)**

- Small gaps exist between the end of one neuron, i.e. the **axon terminals**, and the beginning of the next neuron, i.e. the **dendrites**.
- These gaps are called **synapses** or **synaptic clefts**.
- Electric currents cannot pass across these gaps, so special chemicals are released into the synaptic cleft.
- These chemicals are contained in vesicles in the axon terminals and are called **neurotransmitters**.
- Neurotransmitters were discovered by the German pharmacologist, Otto Loewi on work he conducted on freshly killed frogs.

- Neurotransmitters move across the synaptic cleft by diffusion and bind onto receptors on the dendrite of the next neuron. This causes sodium gates to open, setting up an action potential in the next neuron.
- Neurotransmitters include acetylcholine and noradrenaline.

*Extra information about how synaptic transmission occurs*

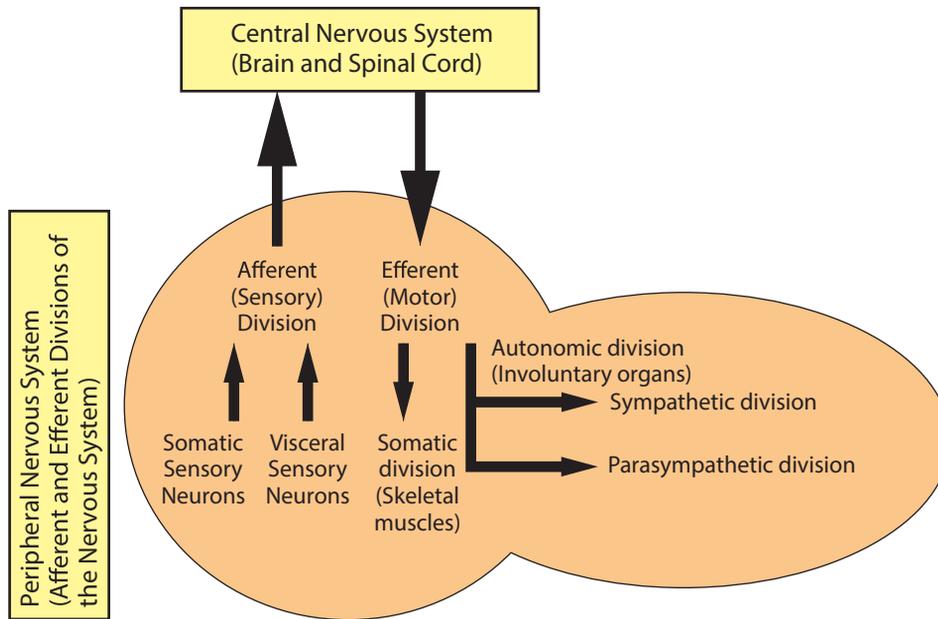
- The arrival of the impulse at the axon terminal causes calcium ion gates to open.
- Calcium ( $\text{Ca}^{2+}$ ) enters the axon terminal through these gates by facilitated diffusion.
- The influx of calcium causes the vesicles containing the neurotransmitter to bind with the plasma (cell surface) membrane. The neurotransmitter is released into the synaptic cleft by exocytosis.
- The neurotransmitter diffuses across the membrane binding with the specific receptor proteins for that specific neurotransmitter on the surface of the dendrite.
- This alters the permeability of the dendrite to sodium, by causing sodium ion gates to open, thereby setting off an action potential in this dendrite.
- An enzyme specific to the neurotransmitter is released and this breaks down the neurotransmitter.
- The broken down neurotransmitter leaves the receptor sites and diffuses back across the synaptic cleft.
- It re-enters the axon terminal by endocytosis and is reformed into the neurotransmitter, back in the vesicles and ready to be used again.

## THE ORGANISATION OF THE NERVOUS SYSTEM

- The nervous system is made up of two main parts; the central nervous system and the peripheral nervous system.
- **The central nervous system (CNS):** Consists of the brain and the spinal cord.
- **The peripheral nervous system (PNS):** Carries messages to and from the CNS from the receptors to the effectors. The 12 pairs of nerves that derive from the brain are called **cranial** nerves, whereas the 31 pairs of nerves derived from the spinal cord are called **spinal** nerves. Within the pair, one nerve leads to the CNS (**sensory fibres**) and one leads away (**motor fibres**).

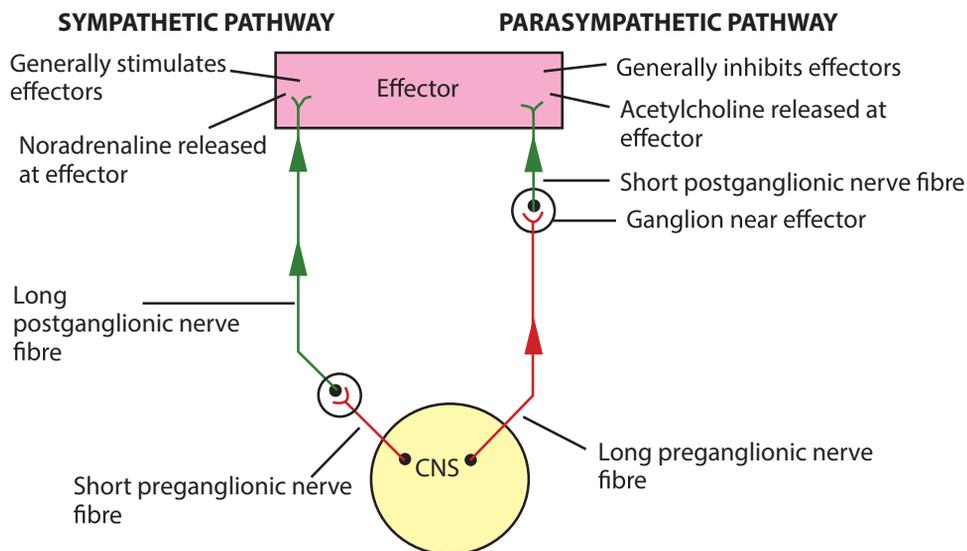
The PNS is divided into two main parts:

- o **The afferent (or sensory) division:** Carries nerve impulses to the CNS.
  - **Somatic sensory neurons:** Carry messages from the skin and muscles.
  - **Visceral sensory neurons:** Carry messages from internal organs.
- o **The efferent (or motor) division:** Carries nerve impulses away from the CNS. It is subdivided into:
  - **The somatic division:** Takes nerve impulses from the CNS to the skeletal muscles.
  - **The autonomic division:** Takes nerve impulses from the CNS to the heart muscle, involuntary muscles and glands. It is further divided into:
    - **The sympathetic division:** Tends to produce responses that prepare the body for action.
    - **The parasympathetic division:** Tends to maintain the body during normal activity.



### THE AUTONOMIC DIVISION OF THE NERVOUS SYSTEM (ANS)

- *Auto* – means ‘self’, *nomo* – means ‘govern’. Therefore, the autonomic system is self-governing.
- It usually controls the involuntary activities of smooth muscle and certain glands, whereas the somatic division is usually under voluntary control, i.e. the skeletal muscles.
- Unlike the skeletal muscles which has one nerve fibre leading from the CNS, each pathway within the ANS consists of a **preganglionic neuron** and a **postganglionic neuron**. A ganglia is a group of cell bodies located outside of the CNS. This means that there are two nerve fibres in the ANS nerve fibres, with a synapse between them. (See diagram below)
- The neurotransmitter released at the effector from the somatic pathway is always acetylcholine. This is not the case for the ANS. In the sympathetic division, **noradrenaline** is released, whereas in the parasympathetic pathway **acetylcholine** is released.



- The sympathetic and parasympathetic pathways normally work together but with opposing actions, i.e. they are **antagonistic**. Generally the sympathetic pathway prepares the body for action and the parasympathetic pathway works to keep the body at rest. However, it isn't true to say that one pathway speeds up organ functioning and the other slows it down.

- In threatening or stressful situations, the body responds by preparing the body for action. This usually activates the sympathetic division and can result in a **fight-or-flight response (alarm reaction)**.
- A comparison between the sympathetic and parasympathetic nervous system can be seen below:

| Structure(s)        | Sympathetic nervous division                                      | Parasympathetic nervous division                                    |
|---------------------|-------------------------------------------------------------------|---------------------------------------------------------------------|
| Heart               | Increases cardiac output                                          | Decreases cardiac output                                            |
| Eyes                | Dilates pupils                                                    | Constricts pupils                                                   |
| Lungs               | Dilates bronchioles                                               | Constricts bronchioles                                              |
| Lungs               | Increases breathing rate                                          | Decreases breathing rate                                            |
| Bladder and anus    | Contracts anal and bladder sphincters                             | Relaxes anal and bladder sphincters                                 |
| Salivary glands     | Decreases saliva production (dry mouth)                           | Increases saliva production                                         |
| Liver/gall bladder  | Increases breakdown of glycogen to glucose/ inhibits gall bladder | Increases conversion of glucose to glycogen/stimulates gall bladder |
| Stomach, intestines | Inhibited movement                                                | Stimulates movement                                                 |
| Adrenal medulla     | Stimulates hormone secretion                                      | No obvious effect                                                   |
| Sweat glands        | Greater sweat production                                          | No obvious effect                                                   |
| Skin                | Vasoconstriction of blood vessels                                 | Little effect                                                       |
| Skeletal muscles    | Vasodilation of blood vessels                                     | No effect                                                           |
| Internal organs     | Vasoconstriction of blood vessels of all except heart and lungs   | Little effect                                                       |

### Differences between the nervous and endocrine systems

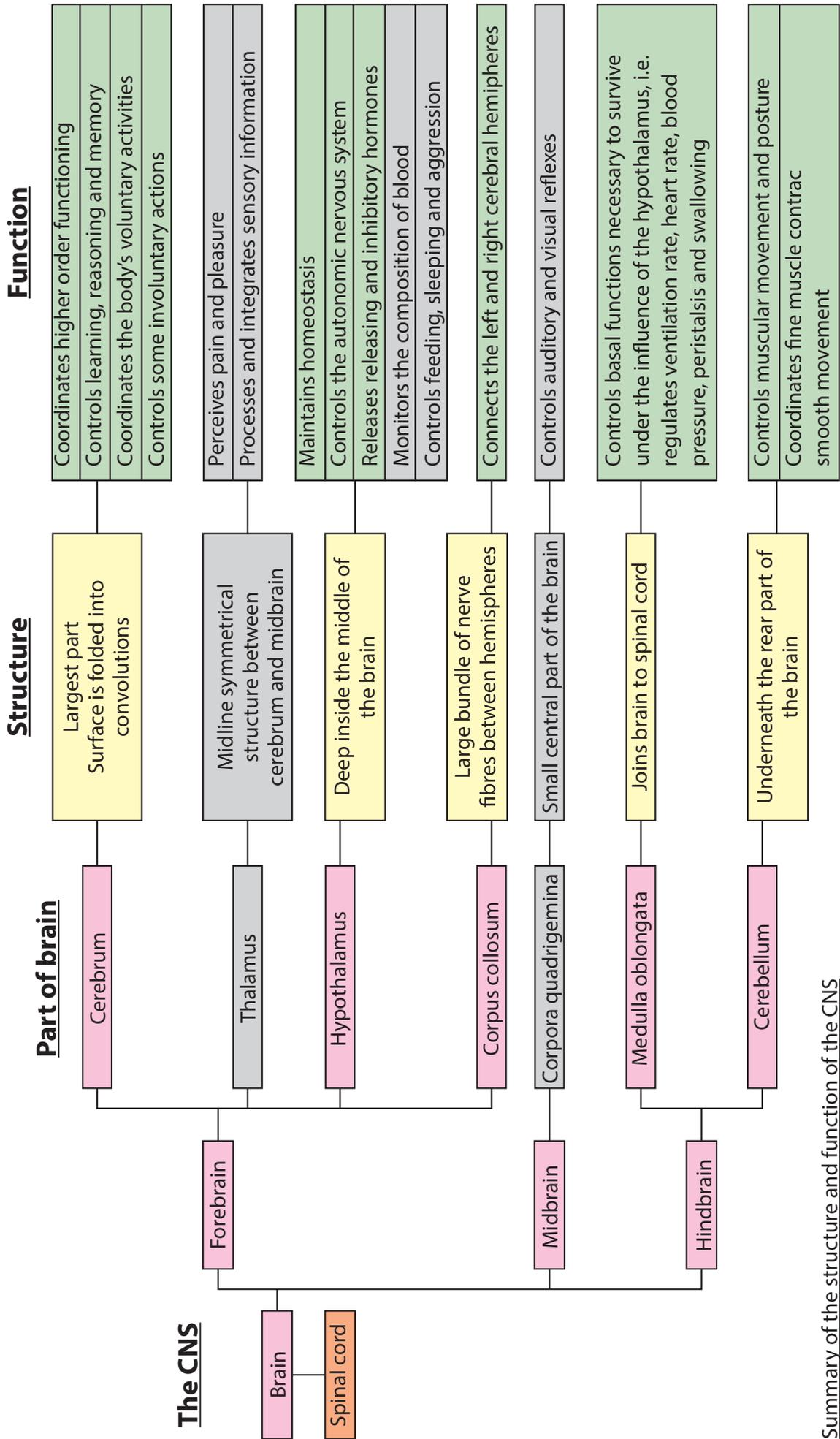
- Whilst the nervous and endocrine systems often work together, they do have differences. These can be seen in the table below:

|                    | Nervous system                                              | Endocrine system                                        |
|--------------------|-------------------------------------------------------------|---------------------------------------------------------|
| Speed of action    | Rapid/Fast – within milliseconds                            | Slower – from seconds to days                           |
| Duration of action | Brief – usually short-lived                                 | Longer lasting – usually after the stimulus has stopped |
| Nature             | Electrochemical (Electrical impulses and neurotransmitters) | Chemical only (Hormones)                                |
| Transmission       | Along the membranes of neurons/nerve fibres                 | Through the bloodstream                                 |
| Specificity        | Effects are localised                                       | Effects are usually widespread and general              |

### The Central Nervous System

- The CNS is made up of the **brain** and the **spinal cord**.
- A summary of the structure and function of the central nervous system (CNS) can be seen on the next page:





Summary of the structure and function of the CNS  
(Parts in grey are not in the syllabus)

### a. Protection of the CNS

- The brain and the spinal cord are vital organs and need to be protected.
- Three levels of protection exist:

#### 1. Bone:

- The brain is protected by the **cranium** (Exam hint: Not the skull. The skull includes the jawbone/mandible, which doesn't protect the brain). Thus, the bones of the skull help protect the brain.
- The spinal cord is protected by the **vertebrae**, which together form the **vertebral canal**.
- Both the cranium and the vertebral canal are hard layers, which are difficult to penetrate

#### 2. Membranes called meninges:

- These cover the entire CNS and are made up of tough, fibrous tissue.
- There are three meninges, which provide a tough, fibrous protective layer.

- **Dura mater:** Thick durable membrane. It is attached to the skull, but not to the vertebral canal. Instead, in the vertebral canal, there is a space filled with fat, connective tissue and blood vessels. The dura mater is a fibroelastic layer of cells. It contains larger blood vessels that split into capillaries in the pia mater.
- **Arachnoid mater:** It has a spider web-like appearance, hence its name. It cushions the CNS. It is a thin, transparent membrane, which is thought to be impermeable to fluid.
- **Pia mater:** This layer firmly adheres to the surface of the brain and spinal cord, much like a fibrous glove. Like the dura mater, it is thought to be impermeable to fluid. It is a very thin layer of fibrous tissue, which is punctuated by blood capillaries to the brain and spinal cord. These allow the CNS to be nourished.

- An inflammation of these membranes is called **meningitis**, which can be caused by a virus or bacteria. This can place dangerous pressure on the brain.

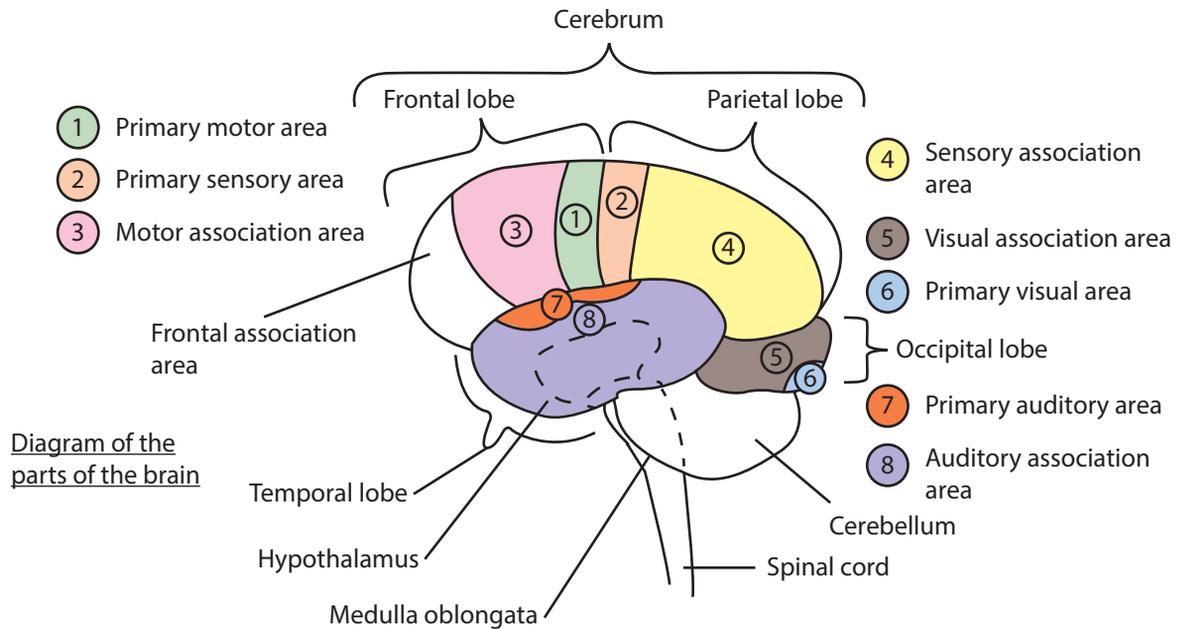
#### 3. Cerebrospinal fluid (CSF):

- This clear, watery fluid contains a few cells, some glucose, protein, urea and salts, and occupies the space between the middle and inner layers of the meninges.
- It circulates through the ventricles (cavities) of the brain, providing nutrients to the brain and spinal cord, whilst also removing waste products.
- CSF is formulated from blood and, after it circulates around and through the CNS, it returns to blood capillaries.
- Also, the CSF acts as a shock absorber, cushioning the CNS from any blows or shocks it may receive.

### b. Structure of the CNS

- Much of the structure of the brain can be seen in the summary diagram, together with its function.

**Exam hint:** You don't have to be an artist to draw a diagram of the main lobes of the cerebrum. See next page.



- **Cerebrum**

- o Biggest part of the brain.
- o Outer surface is about 2-4 mm thick, made up grey matter and is called the **cerebral cortex**.
- o In the deepest parts of the cerebrum is grey matter called **basal ganglia** (Ganglia is the name given to a group of nerve cell bodies).
- o Cerebrum is divided into two hemispheres (left and right). The halves are joined by a network of nerve fibres, called the **corpus callosum**. These fibres allow communication between the two hemispheres.
- o The cerebrum is folded to increase its surface area. These folds are known as **convolutions** or gyri (singular: gyrus (Think: gyrating, which means to move from side to side, although, technically, these are raised ridges on the cerebrum)). Between these folds are grooves, which are either shallow, called **sulci** (singular: **sulcus**), or deep, called **fissures**. The deepest fissure, which houses the corpus callosum, is between the two hemispheres and is called the **longitudinal fissure**.
- o Cerebral cortex and basal ganglia are bundles of nerve fibres called **tracts**, which are myelinated and so sometimes this is referred to as white matter. Tracts connect various areas of the cortex within the same hemisphere, they carry impulses between the left and right hemispheres, and they connect the cortex to other parts of the brain or to the spinal cord.
- o Within the cortex are three main functional areas that allow for thinking, reasoning, learning, memory, intelligence, as well as coordinating the body's voluntary activities in response to the senses received.
  - **Sensory areas:** Receive and process nerve impulses from the afferent pathway.
  - **Motor areas:** Send impulses to effectors via the efferent pathway, e.g. skeletal muscles.
  - **Association areas:** Interpret information from the senses and (usually in light of previous experience) make it useful.

- **Cerebellum**

- o Positioned under the rear part of the cerebrum.
- o It is the second largest part of the brain and its surface is folded into a series of parallel ridges.

- o It is a large and complex association area concerned with the control of fine muscular movement and body posture.
- o It receives sensory information from the skeletal muscles and stretch receptors in the inner ear.
- o Its role is **not** to initiate movement, but to coordinate it.
- o Any damage to the cerebellum results in jerky and uncontrolled movement.
- **Hypothalamus**
  - o The main controlling region for the autonomic nervous system.
  - o Although small, it controls many functions, including such complex patterns of behaviour as feeding, sleeping and aggression, but it is mostly concerned with homeostasis.
- **Medulla oblongata**
  - o A continuation of the spinal cord.
  - o Contains the control centres of the most basic functions necessary for survival.
    - **Cardiac centre:** Regulates the rate and force of heart beat.
    - **Respiratory centre:** Regulates rate and depth of breathing.
    - **Vasomotor centre:** Regulates the diameter of blood vessels.
  - o All the control centres of the medulla oblongata are influenced and regulated by higher centres of the brain.
  - o Plays an important role in automatically adjusting body functions.
- **Spinal cord**
  - o A dorsal (at the back) cylinder of nervous tissue running from the bottom of the brain, passing through the opening called the **foramen magnum** at the base of the skull and travelling the length of the body to the second lumbar vertebra.
  - o The vertebrae of the vertebral canal provide protection to it, as does the fatty tissue, the meninges and CSF. (See earlier notes).
  - o A cross-section through the spinal cord reveals two sections:
    - **Grey matter:** composed of cell bodies and unmyelinated nerve fibres and found at the centre.

**Exam hint:** When asked about this in an exam, both parts of the composition are needed, i.e. the underlined word 'and' is important.

- **White matter:** composed of myelinated fibres.

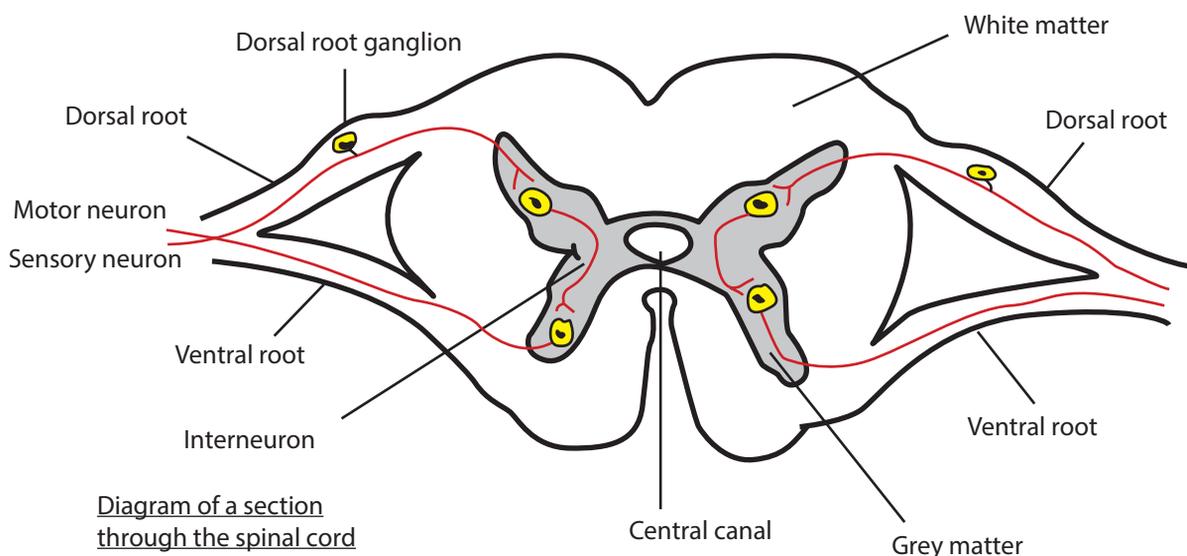


Diagram of a section through the spinal cord

- o **Central canal:** Contains ascending and descending tracts.
  - **Ascending tracts:** Sensory/afferent neurons carry impulses up the spinal cord towards the brain.
  - **Descending tracts:** Motor/efferent neurons carry impulses down the spinal cord away from the brain.
- o **Dorsal root** (uppermost): Carries sensory/afferent neurons only.
- o **Ventral root** (lower): Carries motor/efferent neurons only.
- o **Dorsal root ganglia:** Is a swelling in the dorsal root, which houses the cell bodies of sensory neurons.

### c. Reflexes

- The spinal cord integrates certain reflexes.
- A reflex is a rapid, automatic response to a change in the external or internal environment.
- Protective reflexes, as their name suggests, are designed to remove a person from harm.
- It is **incorrect** to say that reflexes “don’t involve the brain”. It is correct to say that the response is **unconscious** in the sense that the reflex occurs before the information reaches the brain in the ascending tract and the brain plays no part in the response.
- Reflexes share certain characteristics:
  - o A sensory **stimulus** is needed to activate the reflex, i.e. it is not spontaneous.
  - o It is **involuntary**. No conscious thought is involved.
  - o It is **rapid**. Only a small number of neurons are involved, with usually only one or two synapses between the stimulus and response.
  - o It is **stereotypical**. It occurs the same way every time.
- Any reflex that is localised within the spinal cord is called a **spinal reflex**.
- The pathway of neurons involved in the reflex action is known as a **reflex arc**. The pattern is as follows:
  - o A stimulus is detected by **receptor** cells, e.g. pain receptors pick up the stimulus of a pin in the skin.
  - o The **sensory/afferent neuron** carries the impulse from the receptor to the spinal cord.
  - o The impulse either passes directly to a **motor/effector neuron** across at least one **synapse** or via an interneuron.
  - o The motor/effector neuron carries the message to the effector (either a muscle or gland).
  - o The effector, in this case a muscle, which contracts, carries out a **response**.
- Some reflexes are not protective, but are brought about by the autonomic nervous system, e.g. production of saliva in response to food, ejaculation of semen during sexual intercourse.
- **Learned responses** are sometimes referred to as **learned reflexes**, **acquired reflexes** or **conditioned responses/reflexes**.
  - o These responses are as a result of repetition, resulting in the development of seemingly an unconscious response, i.e. thinking doesn’t seem to be involved in the process.
  - o Often these responses develop as a result of two stimuli being presented together, e.g. the school bell rings signalling the beginning of lunch results in salivation occurring.
  - o These learned responses can be reinforced by repetition, but do weaken if they aren’t reinforced, even if they aren’t lost altogether.

## Types of Receptors

- A receptor is a structure that detects a change in the body's internal or external environment.
- When receptor cells of a particular type are grouped together, they form a **sense organ**.
- There are different types of receptors:
  - **Thermoreceptors:** These detect changes to heat or cold. In the skin and mucous membranes, there are two types; heat and cold receptors, whereas thermoreceptors in the hypothalamus can respond to either heat or cold. The core temperature is detected in the thermoregulatory centre of the hypothalamus that stimulates a response to changes accordingly.
  - **Osmoreceptors:** These are found primarily in the hypothalamus and respond to changes in the osmotic pressure. If the concentration of substrates dissolved in the blood is greater than expected, it means there is less water in the blood and the osmotic pressure has increased. This is detected by the osmoreceptors, which stimulate a response to ensure more water is reabsorbed.
  - **Chemoreceptors:** These respond to particular chemicals. They are found in the mouth and nose, allowing us to respond to different tastes and odours respectively. Internal chemoreceptors respond to changes in pH and changes in concentrations of carbon dioxide and oxygen, as well as the composition of body fluids.
  - **Cutaneous receptors:** These are found in the skin and include touch receptors (mechanoreceptors) and pain receptors (nociceptors) and some thermoreceptors (see notes above).
    - **Touch receptors:** Some are close to the skin surface (in the epidermis) and respond to light touches. Others are located much deeper in the skin (in the dermis) and respond to pressure and vibrations. Still others are attached to the base of hair follicles and respond to touches that bend the hair.
    - **Pain receptors:** These are stimulated by a number of factors that result in damage or potential damage, such as damage to tissues or by excessive stimulation, such as stimuli from heat or chemicals. There are internal pain receptors associated with most organs, except the brain, but the majority are found in the skin. Pain can serve as a warning sign to the body, allowing us to recognise that damage may have occurred to a part of our body, or alerting us to seek medical help. This is an advantage of being able to respond to pain.





# QUESTIONS

30 marks

1. State two differences between sensory and motor neurons. [2]

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2. Johnathan was working on his car when he badly gashed his arm. He was still able to move his arm, but couldn't feel any sensation in it. [7]

a. Explain why he was unable to feel his arm, but was still able to move it. (2)

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b. Several neurons are involved in transmitting the signal to Johnathan's arm. Explain how information is transmitted from one neuron to the next. (5)

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3. Complete the table below with reference to the differences between the nervous and endocrine systems. [8]

|                                          | <b>Nervous System</b> | <b>Endocrine System</b> |
|------------------------------------------|-----------------------|-------------------------|
| Nature of transmission                   |                       |                         |
| Duration of action                       |                       |                         |
| Comparative speed                        |                       |                         |
| Overall effect of signal on target organ |                       |                         |

4. State three structures that protect the brain from damage. [3]

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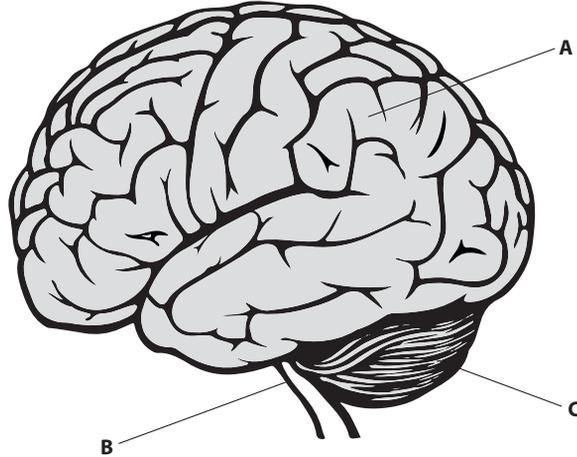


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5. Below is a diagram of the brain: [6]

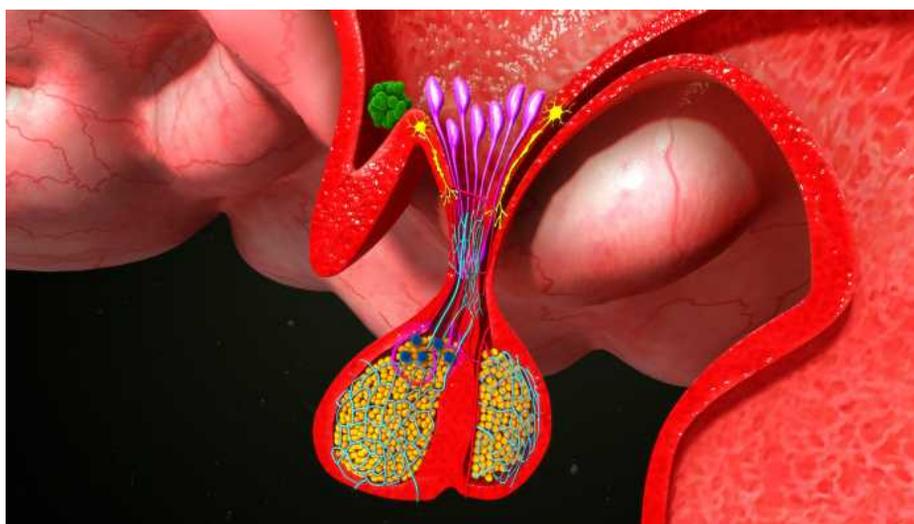


- a. Label parts B and C. (2)
- 
- 
- b. Which letter represents where the vasomotor centre is located? (1)
- 
- c. Which letter represents where decisions are made to carry out writing? (1)
- 
- d. In part A, what are the folds called and what is their significance? (2)
- 
- 
- 
6. Letitia was just about to step off the pavement onto the road, when she heard a screech of brakes. Immediately, she stepped back onto the pavement. [4]
- a. What type of response is this? (1)
- 
- b. Explain how this response is able to protect her from harm. (2)
- 
- 
- c. State one other feature of this type of response that is likely to occur if something similar happens again. (1)
-



# HOMEOSTASIS AND THE ENDOCRINE SYSTEM (UNIT 3)

# 3



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                 | Done | Revise | Teach others |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Chemical Messengers and the Endocrine system (Unit 3)</b>                                                                                                                                                                                                                                                                        |      |        |              |
| <ul style="list-style-type: none"> <li>I can describe the role of the hypothalamus, pituitary, thyroid, parathyroid, pancreas, thymus, gonads, pineal and adrenal glands as endocrine glands found in the human body.</li> </ul>                                                                                                    |      |        |              |
| <ul style="list-style-type: none"> <li>I can describe how hormones secreted from the hypothalamus, pituitary, thyroid, parathyroid, pancreas and adrenal glands are involved in homeostasis by affecting specific target organs.</li> </ul>                                                                                         |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain that hormones secreted from the hypothalamus, pituitary, thyroid, parathyroid, pancreas and adrenal glands are involved in homeostasis by affecting specific target organs.</li> </ul>                                                                                         |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how the secretions of the pituitary gland are controlled by the hypothalamus through transport of hormones, either via nerve cells or the vascular link between them.</li> </ul>                                                                                               |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how hormones can be lipid-soluble and able to cross cell membranes to bind with and activate intracellular receptors or, water-soluble and able to bind with and activate receptors on cell membranes, and require secondary messengers to affect cell functioning.</li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can discuss how synthetic hormones may be developed to control or treat endocrine dysfunction, including diabetes mellitus, hypothyroidism and hyperthyroidism, to improve the quality of life for individuals.</li> </ul>                                                                 |      |        |              |
| <b>Homeostasis: (Unit 3)</b>                                                                                                                                                                                                                                                                                                        |      |        |              |
| <ul style="list-style-type: none"> <li>I understand that homeostatic processes involve nerves and hormones in maintaining the body's internal environment within tolerance limits through the control of metabolism and physiological and behavioural activities.</li> </ul>                                                        |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how thermoregulation occurs by the control of heat exchange and metabolic activity through physiological and behavioural mechanisms.</li> </ul>                                                                                                                                |      |        |              |

| Key teaching points                                                                                                                                                                                                                                                                                      | Done | Revise | Teach others |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <ul style="list-style-type: none"> <li>I can explain how body fluid concentrations are maintained by balancing water and salts via the skin, digestive system and the kidneys, which involve the actions of antidiuretic hormone (ADH) and aldosterone on the nephron, and the thirst reflex.</li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how blood sugar levels are maintained by controlling of sugar uptake, its storage and release by cells and use in metabolism; these processes involve the hormones of the pancreas and adrenal glands.</li> </ul>                                   |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain that concentrations of gases are controlled by balancing the intake of oxygen and the removal of carbon dioxide via the lungs, through the actions of the medulla oblongata and the autonomic nervous system.</li> </ul>                            |      |        |              |
| <ul style="list-style-type: none"> <li>I can appreciate that synthetic hormones may be developed to control or treat endocrine dysfunction, including diabetes mellitus, hypothyroidism and hyperthyroidism, to improve the quality of life for individuals. (SHE)</li> </ul>                            |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how blood sugar levels are maintained by controlling of sugar uptake, its storage and release by cells and use in metabolism and that these processes involve the hormones of the pancreas and adrenal glands.</li> </ul>                           |      |        |              |

### Helpful hints:

- Usually there will be an extended question on this section.
- Ensure you identify the area of homeostasis that the question is asking about. Don't answer everything you know about a specific area of homeostatic control, but only what you're asked. For example, if you are asked about overheating, don't write about overcooling as well.
- Learn the difference between the action of lipid-soluble and water-soluble hormones. It is a very common exam question. Note use in the syllabus of lipid-soluble or water-soluble, not steroid or protein/amine.
- Learn about the relationship between the hypothalamus and the anterior lobe and posterior lobe of the pituitary gland, and the differences between the two lobes.
- Follow the Stimulus → Receptor → Modulator → Effector → Response → Feedback model for your answers (SRMERF)

### Overview:

- Homeostasis** literally means: 'Homo-' (Same), '-stasis' (to keep constant, to maintain). To maintain a constant, internal environment or steady state.
- Homeostasis is necessary, because enzymes work at specific pH, temperature and ion concentration.
- Hormones** are involved in homeostasis. Hormones are **chemical messengers** secreted into the blood plasma by endocrine glands and affect target organs.
- Hormones are released from ductless glands, called **endocrine glands**. These chemical transmitters, which are released into the blood, affect target organs.
- There are two types of gland:

| Exocrine glands                                                                                                         | Endocrine glands                                                                                                       |
|-------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------|
| <ul style="list-style-type: none"> <li>Have ducts and secrete fluids into body cavities or to body surfaces.</li> </ul> | <ul style="list-style-type: none"> <li>Ductless. Secrete hormone into extracellular fluid surrounding gland</li> </ul> |

- There are two types of feedback:

| Positive                                                                                                         | Negative                                                                                                                                     |
|------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------|
| <ul style="list-style-type: none"> <li>Feedback that reinforces or amplifies the original stimulus.</li> </ul>   | <ul style="list-style-type: none"> <li>Feedback that reduces the effect or eliminates the original stimulus.</li> </ul>                      |
| <ul style="list-style-type: none"> <li>Not involved in homeostasis.</li> </ul>                                   | <ul style="list-style-type: none"> <li>Involved in homeostasis.</li> </ul>                                                                   |
| <ul style="list-style-type: none"> <li>Examples: Fever, blood clotting, childbirth and breastfeeding.</li> </ul> | <ul style="list-style-type: none"> <li>Examples: Regulation of blood sugar, gas concentrations, osmoregulation, thermoregulation.</li> </ul> |

- In homeostasis, negative feedback or steady state control systems operate and follow the pattern below:
  - **Stimulus:** Change to the steady state.
  - **Receptor:** Detects the change.
  - **Modulator:** Control centre processes the information and responds, often by stimulating an endocrine gland to release a hormone. (Often this will be the **hypothalamus**).
  - **Effector:** Hormone, or otherwise, counteracts the effect of the stimulus.
  - **Response:** The action of the effector.
  - **Feedback:** Steady state has been restored so system returns to normal.
- **N.B. Both the nervous system and endocrine system are involved in maintaining steady states.**

## HORMONES

- Hormones **don't start reactions** within cells, but **regulate reactions**.
- They're required in minute quantities, can be transferred from one animal to another safely and can't be stored (as the liver will break them down → effects are short-lived).
- **Hypo-** = too little hormone is secreted.
- **Hyper-** = too much hormone is secreted.
- There are two main types: Protein and amine hormones or steroid hormones

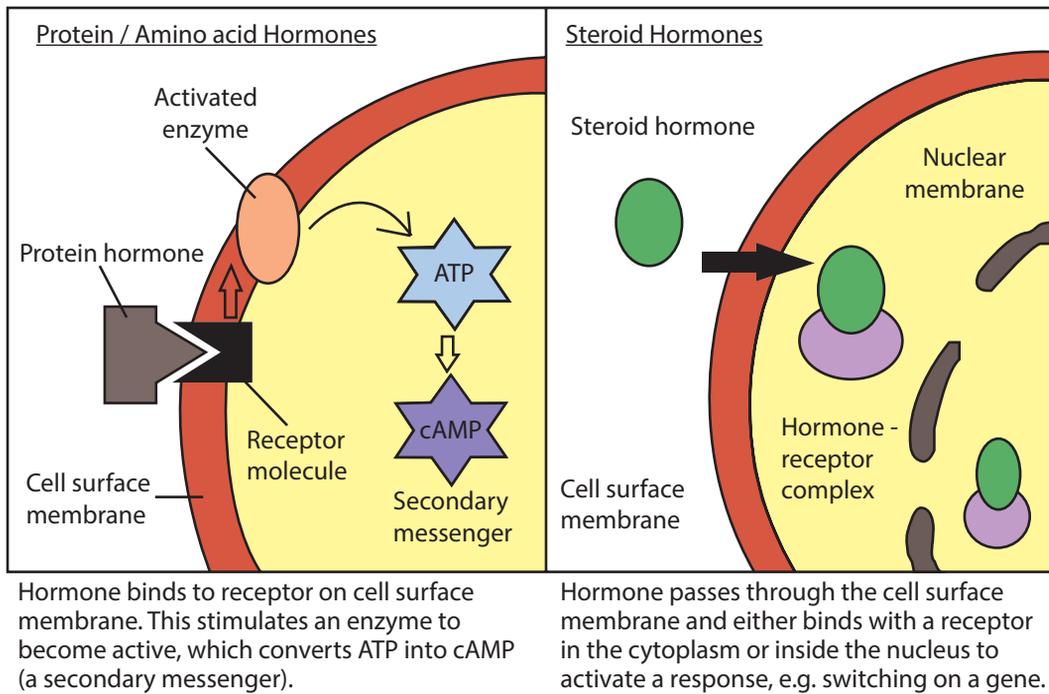
| Water-soluble (Protein/amine)                                                                                                                    | Lipid-soluble (Steroid)                                                                                                                    |
|--------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------|
| <ul style="list-style-type: none"> <li>• Attached to receptor proteins on the cell surface membrane (plasma membrane).</li> </ul>                | <ul style="list-style-type: none"> <li>• Pass across the membrane (non-polar) and bind with a receptor protein inside the cell.</li> </ul> |
| <ul style="list-style-type: none"> <li>• Activate secondary messenger within the cell (usually via activating enzymes – see diagram).</li> </ul> | <ul style="list-style-type: none"> <li>• Activate genes controlling the formation of specific proteins.</li> </ul>                         |

- Hormones are able to change how cells function. This can be done in a number of different ways. For example, by changing the type or quantities of proteins produced, or activities of those proteins.
- Whilst not enzymes themselves, hormones are capable of changing the concentration of enzymes or enzyme activity itself.
- Some of the actions of hormones include:
  - Switching on genes by binding with a repressor protein rendering it inactive. This results in the transcription and translation of structural genes to form proteins/enzymes.
  - Binding to enzymes, causing their shape to change (denaturation), so they are effectively turned off.
  - Changing the rate of production of enzymes.

### *Extra information*

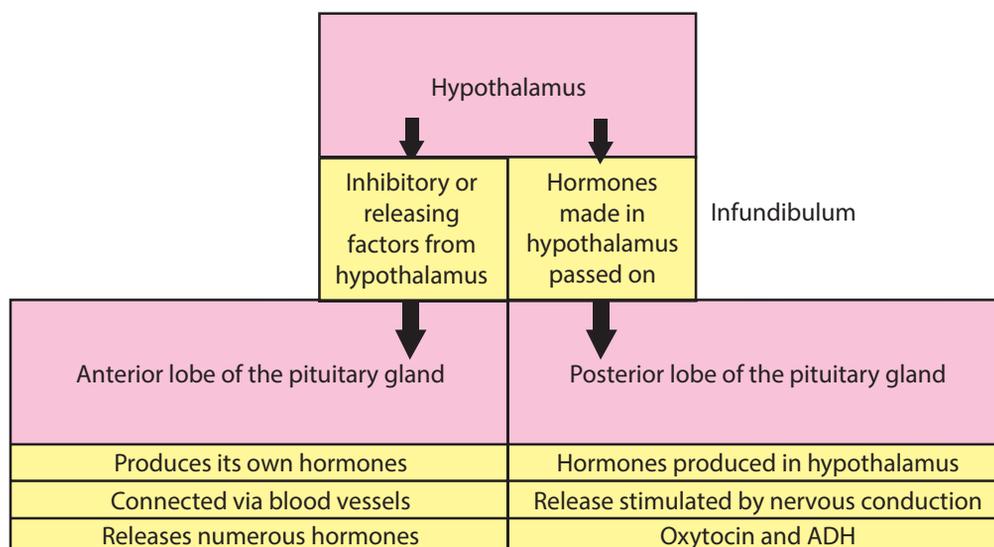
- When cells conduct cell signalling within the same tissue, they do so using localised hormones called **paracrines**. Paracrines move through the extracellular fluid.
- **Enzyme amplification** occurs when a hormone causes the activation of a number of enzymes at the same time. This results in a number of molecules being activated at the same time.
- **Hormone clearance** occurs when the hormone that has caused a specific reaction needs to be deactivated. Usually this is done by enzymes in the target organs. The degraded hormones then pass to the liver to be deaminated or broken down further before passing out through the kidney.

- Below is a diagram of the two types of hormone action:



### THE RELATIONSHIP BETWEEN THE HYPOTHALAMUS AND THE PITUITARY GLAND

- The hypothalamus is found at the base of the brain and is responsible for the regulation of a number of homeostatic mechanisms.
- It regulates activities, such as sleep, thirst and temperature control.
- It monitors levels of hormones and other chemicals in the blood passing through it.
- It is linked to the pituitary gland via the **infundibulum**.
- The pituitary gland is made up of two parts; the **anterior lobe** (front lobe) and **posterior lobe** (rear lobe). These two lobes respond differently and are under the control of the hypothalamus in different ways. (See diagram below):



- The posterior lobe of the pituitary gland (PLPG) is often considered a 'false gland' as it doesn't produce hormones itself, but stores and releases them. The hormones, oxytocin and ADH (or **vasopressin**) are made in special nerve cells in the hypothalamus, which then extend into the PLPG. Release is via nervous stimulation from the hypothalamus.

- The anterior lobe of the pituitary gland (ALPG) releases several hormones as a result of stimulation via tiny blood vessels of releasing factors from the hypothalamus. All the hormones released from the ALPG stimulate the activity of a specific target organ, except for growth hormone, which affects body tissues in general and is secreted throughout life.
- The hormones released from the ALPG and PLPG can be seen in the table below:

| Hormone                                                                 | Target organ                           | Main Effects                                                                                                                                                                                                               |
|-------------------------------------------------------------------------|----------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Anterior Lobe</b>                                                    |                                        |                                                                                                                                                                                                                            |
| Thyroid stimulating hormone (TSH)                                       | Thyroid gland                          | <ul style="list-style-type: none"> <li>• Stimulates growth of the thyroid gland.</li> <li>• Stimulates the growth and release of hormones from the thyroid gland.</li> </ul>                                               |
| Adrenocorticotrophic hormone (ACTH)                                     | Adrenal cortex                         | <ul style="list-style-type: none"> <li>• Regulates growth of the adrenal cortex.</li> <li>• Stimulates the release of hormones from the adrenal cortex.</li> </ul>                                                         |
| Follicle stimulating hormone (FSH)                                      | Ovaries or Testes                      | <ul style="list-style-type: none"> <li>• Initiates maturation of follicles in female ovaries.</li> <li>• Initiates production of sperm in male testes.</li> </ul>                                                          |
| Luteinising hormone (LH) (Interstitial cell stimulating hormone) (ICSH) | Ovaries (in females) Testes (in males) | <ul style="list-style-type: none"> <li>• Causes ovulation and the consequent development of the corpus luteum, which it maintains.</li> <li>• Stimulates the secretion of testosterone.</li> </ul>                         |
| Prolactin (PRL)                                                         | Mammary glands                         | <ul style="list-style-type: none"> <li>• Maintains progesterone production from the corpus luteum.</li> <li>• Induces milk production in pregnant females.</li> </ul>                                                      |
| Growth hormone (GH)                                                     | All cells                              | <ul style="list-style-type: none"> <li>• Promotes growth of skeleton and muscles.</li> <li>• Controls protein synthesis and general body metabolism.</li> </ul>                                                            |
| <b>Posterior Lobe</b>                                                   |                                        |                                                                                                                                                                                                                            |
| Antidiuretic hormone (ADH)                                              | Kidneys                                | <ul style="list-style-type: none"> <li>• Causes an increase in the permeability of the distal convoluted tubules and collecting ducts in the nephrons of the kidney, resulting in increased water reabsorption.</li> </ul> |
| Oxytocin (OT)                                                           | Uterus Mammary glands                  | <ul style="list-style-type: none"> <li>• Induces birth by causing uterine contractions.</li> <li>• Induces lactation (secretion of milk from the nipples).</li> </ul>                                                      |

## THE OTHER ENDOCRINE GLANDS

### 1. The pineal gland

- Found deep within the brain and, in children, is about the size of a pea.
- It decreases in size after puberty and its exact function is uncertain.
- It secretes **melatonin**, which is involved in the regulation of sleep patterns.
- Production of melatonin is reduced by being in bright light and levels increase in darkness.
- Some people take it artificially in order to treat insomnia or jet-lag.

### 2. The thyroid glands

- Located in the neck, just below the larynx, consisting of two lobes found either side of the trachea and joined by a narrow piece of tissue.
- It secretes **thyroxine** ( $T_4$ ), its main hormone, as well as **triiodothyronine** ( $T_3$ ) and **calcitonin**.
- Thyroxine and triiodothyronine are very similar chemically and functionally, regulating body metabolism. Both hormones are derivatives of the amino acid tyrosine and both contain iodine.

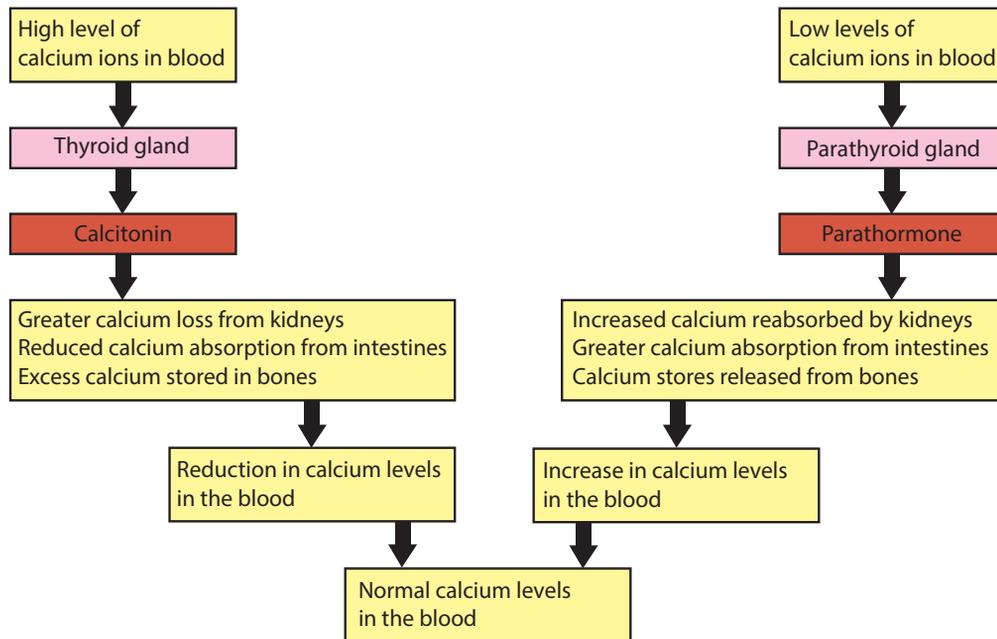
**Extra information**

- *Calcitonin is involved in calcium metabolism. It works in conjunction with parathormone, released from the parathyroid glands. Calcitonin reduces calcium in the blood, whereas parathormone increases calcium in the blood.*

3. The parathyroid glands

- Usually people have four parathyroid glands embedded into the rear surface of the lobes of the thyroid gland.
- They secrete **parathyroid hormone** (or **parathormone**), which increases calcium in the blood, as well as regulating phosphate levels.

A Summary of the Control of Calcium in the Blood

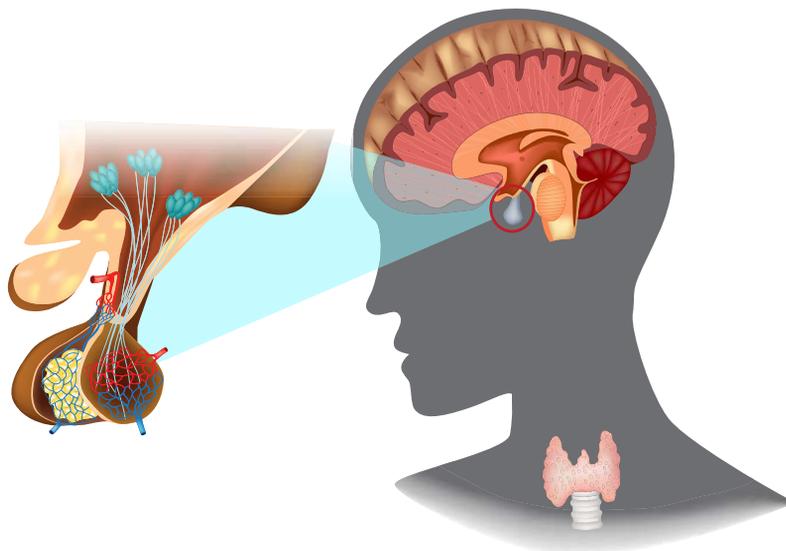
4. The thymus

- Located in the chest, just above the heart and behind the sternum.
- It is larger in infants than in adults and begins to shrink after puberty.
- It secretes **thymosins**, which stimulate the development of disease-fighting T-cells.

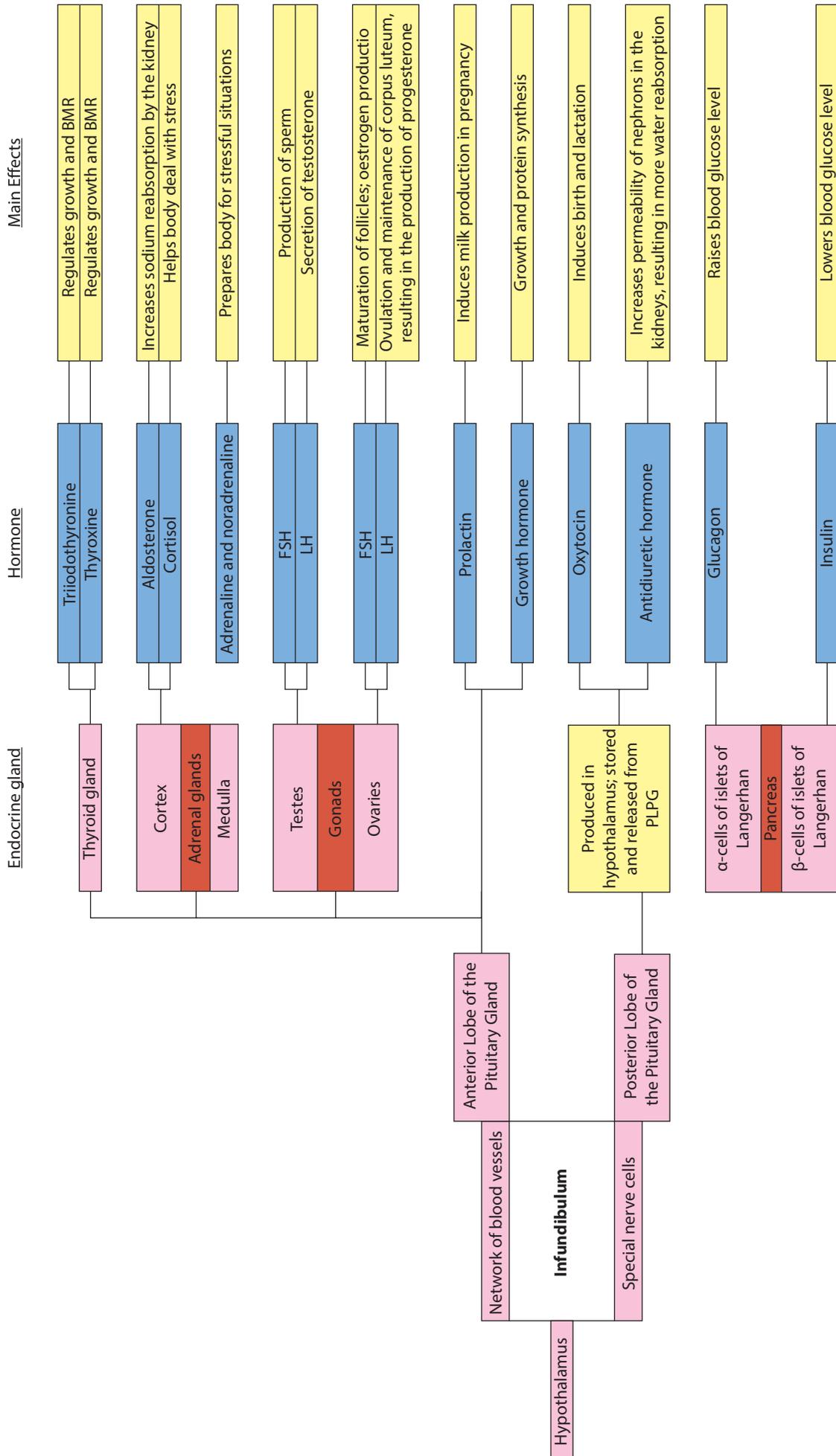
5. The adrenal glands

- These are situated above each kidney.
- They have two separate and independent parts:
  - o **The adrenal cortex**
    - Makes up about 80% of the adrenal gland.
    - Produces a number of hormones which have relatively slow, long-lasting effects on body metabolism.
    - All the hormones produced are **steroids** produced from **cholesterol**.
    - Collectively, hormones from the adrenal cortex are called **corticosteroids** (or **corticoids**), which fall into two groups; **glucocorticoids** (concerned with glucose metabolism) and **mineralocorticoids** (concerned with mineral metabolism).
    - **Cortisol** is one of the glucocorticoids.
      - It is produced in response to stress.
      - In stressful situations, the hypothalamus stimulates the ALPG to release adrenocorticotrophic hormone (ACTH), which stimulates the adrenal cortex to increase the production of glucocorticoids.

- Glucocorticoids combat stress by raising blood sugar levels, partly by inhibiting insulin and partly by gluconeogenesis. Also, glycogen formation is increased in the liver and amino acid uptake by the liver increases.
  - **Aldosterone** is one of the mineralocorticoids.
    - It regulates water retention by acting on the kidneys to regulate the distribution of sodium and other minerals in the tissues. By increasing the reabsorption of sodium and chloride ions, more potassium ions are lost in the urine.
  - o **The adrenal medulla**
    - It produces two hormones; **adrenaline (epinephrine)** and **noradrenaline (norepinephrine)**.
    - Both hormones are involved in preparing the body for action, especially in threatening situations.
    - The cells producing these hormones are effectively modified neurons, and noradrenaline is produced by the sympathetic nervous system.
    - Consequently, these hormones are often seen as the link between the nervous and endocrine systems and their effects are similar to that of the sympathetic division of the nervous system.
6. The pancreas
- It is both an endocrine and exocrine gland.
  - Within the tissue of the pancreas are specialised cells called **islets of Langerhans (or pancreatic islets)**.
  - There are two types:
    - o  $\alpha$ -cells: which secrete the hormone **glucagon** in order to increase blood glucose levels.
    - o  $\beta$ -cells: which secrete the hormone **insulin** in order to decrease blood glucose levels.
7. The gonads
- **Androgens**: Male sex hormones produced by the testes, which are responsible for the development and maintenance of the male sexual characteristics.
  - **Oestrogens and progesterone**: Female sex hormones produced by the ovaries, which stimulate the development and maintenance of the female sexual characteristics, as well as working with FSH and LH to regulate the menstrual cycle, and their involvement in changes that occur during pregnancy.
  - Other organs do secrete hormones, but are not regarded as major endocrine glands.
  - A summary of how the main endocrine glands are linked can be seen on the next page:



# A SUMMARY OF THE ENDOCRINE SYSTEM



## HOMEOSTASIS AND REGULATION

- Homeostatic systems are self-regulating. Therefore, they operate using feedback mechanisms. Once conditions are returned to within **tolerance limits**, i.e. within an upper and lower limit around a **set point** then the regulating mechanism is effectively switched off. Therefore, homeostatic mechanisms are maintained by negative feedback (or **steady state control systems**).
- As stated above, steady state control systems follow the same pattern;

STIMULUS → RECEPTOR → MODULATOR → EFFECTOR → FEEDBACK

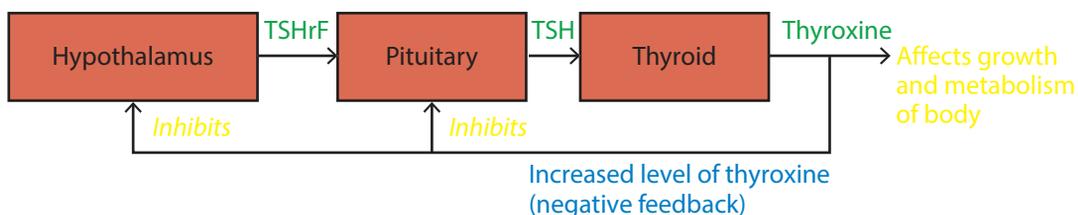
- The following areas will be studied:
  1. Thermoregulation: Control of body temperature.
  2. Osmoregulation: Control of body fluids.
  3. Blood glucose concentration
  4. Regulation of gas concentrations.
- 1. Thermoregulation: Control of body temperature
  - The core human body temperature is about 36.8 °C.
  - Heat production and heat loss is regulated to keep the body as close to this temperature as possible.
  - The optimum temperature for chemical reactions in the body is 37 °C, because this is the temperature that the enzymes work best at. Outside of this range results in either a decrease in the rate of reaction or a denaturation of the enzymes.
  - For the most part, the human body is maintained at a higher temperature than the external environment, because of heat produced as a result of metabolic activity, e.g. when substrates are broken down during aerobic respiration to release energy; some of this energy is released as heat. Heat can be generated by exercise, due to increased muscular activity and, consequently, increased respiration. Additionally, heat may be gained from the external surroundings by conduction and radiation.
  - Heat can be lost from the body as a result of radiation, conduction and convection to the surroundings, and evaporation of water from the skin and lungs.
  - It is vital to keep the body from getting too hot, **overheating**, or too cold, **overcooling**.
  - Changes in temperature are detected by **thermoreceptors**. There are two types; **peripheral**, found in the skin and some mucous membranes and **central**, found in the **thermoregulatory centre** of the **hypothalamus**.
  - The peripheral thermoreceptors consist of two types; **cold receptors**, which are stimulated by decreases in temperature below tolerance levels, and **heat receptors**, which are stimulated by increases in temperature above the tolerance levels.
  - Peripheral thermoreceptors work in conjunction with the central thermoreceptors to ensure that the core temperature remains within the tolerance levels. Other central thermoreceptors exist, which feedback information to the thermoregulatory centre.
  - The hypothalamus regulates temperature in two ways; through nervous conduction to cause physiological and behavioural changes (where possible) to increase or decrease body temperature, and through hormonal control.
- a. Nervous Control
  - Messages are sent via the autonomic nervous system to make physiological changes, mainly in the skin, but also in the muscles.
  - Also, messages are relayed to the cerebrum to coordinate a conscious behavioural response to the conditions, where possible.

|                        | Over-cooling                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       | Over-heating                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            |
|------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Stimulus               | Drop in temperature below tolerance level.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | Increase in temperature above tolerance level.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |
| Receptors              | Cold receptors in skin and mucous membranes.<br>Thermoreceptors in the thermoregulatory centre of the hypothalamus.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Heat receptors in skin and mucous membranes.<br>Thermoreceptors in the thermoregulatory centre of the hypothalamus.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
| Modulator              | Nervous conduction along the sympathetic pathway of the nervous system from the hypothalamus to activate warming processes. Other nervous stimulation occurs to promote shivering.<br>Messages to the cerebrum stimulate behavioural responses.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | Hypothalamus stimulates many physiological changes by nervous conduction, but reduction of nervous conduction from the hypothalamus via the parasympathetic pathway results in vasodilation in the skin. Messages to the cerebrum stimulate behavioural responses.                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| Effectors and response | <ul style="list-style-type: none"> <li>• <b>Vasoconstriction</b> ('vessels narrow') in the skin. This means less blood flows through these capillaries; therefore less heat is lost through radiation, convection and conduction.</li> <li>• <b>Shivering.</b> The rhythmic oscillations of muscles cause tremors, which can generate heat.</li> <li>• <b>Behavioural responses:</b> <ul style="list-style-type: none"> <li>◦ Put more clothes on.</li> <li>◦ Put on a heater.</li> <li>◦ Decrease surface area by curling up into a ball.</li> <li>◦ Increase physical activity.</li> </ul> </li> <li>• <b>(Piloerection.</b> The hair muscles in the skin contract causing the hair to stand up in an attempt to trap a layer of air. This causes the skin to have a pimple like complexion ('Goose-bumps'). It is unlikely that this will keep the body from feeling cold, but the response is to reduce heat loss).</li> </ul> | <ul style="list-style-type: none"> <li>• <b>Vasodilation</b> ('vessels widen') in the skin. This allows for greater blood flow, so the response is more heat is lost by radiation, convection and conduction. (Common misconception: Blood vessels move closer to the surface of the skin. No they don't!)</li> <li>• <b>Sweat glands produce sweat.</b> The evaporation of the sweat from the surface of the skin cools the body down.</li> <li>• <b>Behavioural responses:</b> <ul style="list-style-type: none"> <li>◦ Take clothes off.</li> <li>◦ Turn on air conditioning or a fan.</li> <li>◦ Increase surface area by spreading out.</li> <li>◦ Reduce physical activity.</li> <li>◦ Stand in the shade.</li> </ul> </li> </ul> |
| Feedback               | The temperature increases in the body, unless the external conditions are too great.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | The temperature decreases in the body, unless the external conditions are too great.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |

### b. Hormonal Control

- The adrenal medulla is stimulated when we overcool, resulting in the release of adrenaline and noradrenaline into the blood. This causes an increase in cellular metabolism, which, in turn, generates more heat.
- The main way of generating heat or reducing heat is through the regulation of thyroxine.
- The diagram below summarises how this effect works. Increased thyroxine results in increasing the basal metabolic rate (BMR), resulting in generating increased heat. This is important when overcooling. The effect takes longer to occur, but is longer lasting.
- During overcooling, the hypothalamus produces **thyroid stimulating hormone releasing factor (TSHrF)**, (also called **thyrotropin-releasing hormone (TRH)**) which in turn, stimulates the anterior lobe of the pituitary gland to release more **thyroid stimulating hormone (TSH)**, which then stimulates the thyroid gland to release more thyroxine. This results in an increase in the BMR, leading to warming the body up.
- As the level of thyroxine builds up in the blood it suppresses TSHrF and TSH production. By this form of negative feedback, the level of thyroxine is maintained in the blood.

- The diagram below illustrates how these hormones interact with each other:



c. Thyroxine disorders

- When too much (hyper-) or too little (hypo-) of a hormone is released, it causes problems.
- Hyperthyroidism** occurs when the thyroid gland produces too much thyroxine. The most common type is **Graves’ disease**.
- Hypothyroidism** occurs when the thyroid gland fails to produce enough thyroxine. It is more common than hyperthyroidism, affecting 6-10% of Australian women. In an attempt to secrete more thyroxine in response to TSH release, the thyroid gland becomes inflamed, forming a **goitre**. It is believed as high as 40% or more of the Australian population may suffer from iodine deficiency, albeit to differing degrees, a substance necessary for making both thyroxine and triiodothyronine. Therefore, the federal government introduced iodine supplements into most types of bread in October 2009.

|                      | <b>Hypothyroidism</b>                                                                                                                                                                                                                               | <b>Hyperthyroidism</b>                                                                                                                                                                                                                         |
|----------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Problem              | Too little thyroxine produced.                                                                                                                                                                                                                      | Too much thyroxine produced.                                                                                                                                                                                                                   |
| Symptoms             | <ul style="list-style-type: none"> <li>Slow heart rate.</li> <li>Unexplained weight gain.</li> <li>Fatigue or lacking energy.</li> <li>Intolerance to cold.</li> <li>Swelling to the face.</li> <li>Goitre.</li> </ul>                              | <ul style="list-style-type: none"> <li>Rapid heartbeat.</li> <li>Weight loss.</li> <li>Increased appetite.</li> <li>Fatigue (Extreme tiredness)</li> <li>Sweating.</li> <li>Anxiety.</li> <li>Graves’ disease: protruding eyeballs.</li> </ul> |
| Treatment/Management | <ul style="list-style-type: none"> <li>Extra iodine in the diet, e.g. use of iodised table salt.</li> <li>Tablets containing thyroid hormone. Synthetic forms of these tablets (levothyroxine, which is a form of T4) may be prescribed.</li> </ul> | <ul style="list-style-type: none"> <li>Drugs which block thyroid gland.</li> <li>Surgery to remove part of thyroid gland.</li> <li>Drinking radioactive iodine which targets thyroid cells, which are killed by the radioactivity.</li> </ul>  |

2. Osmoregulation: Control of body fluids

- The amount of water entering the body needs to equal the amount of water leaving it.
- The human body is made up approximately 60% water, contained in various body fluids:

| <b>Type of body fluid</b>                                                                                                                      | <b>Approx. proportion of total body fluid</b>                                                                                                                                                  | <b>Components of the body fluid</b>                                                                                                                                                                                                                                                   |
|------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Intracellular fluid                                                                                                                            | $\frac{2}{3}$ of total body water                                                                                                                                                              | The cytosol (fluid inside the cell)                                                                                                                                                                                                                                                   |
| Extracellular fluid <ul style="list-style-type: none"> <li>Plasma</li> <li>Intercellular fluid (interstitial fluid or tissue fluid)</li> </ul> | $\frac{1}{3}$ of total body water <ul style="list-style-type: none"> <li><math>\frac{1}{4}</math> of extracellular fluid.</li> <li><math>\frac{3}{4}</math> of extracellular fluid.</li> </ul> | Fluid found outside of the cells <ul style="list-style-type: none"> <li>Fluid part of blood.</li> <li>Lymph, cerebrospinal fluid, fluids of eyes and ears fluid in the chest and abdominal cavities and around the heart, fluids of the alimentary canal, kidney filtrate.</li> </ul> |

- Whilst the fluids in each compartment are separated by membranes, they are not isolated from one another. Materials are constantly exchanged between compartments.
- Large molecules, such as plasma proteins, tend to remain inside the fluid in which they are found, because they are generally too big to move across the membrane.
- Water is able to move freely between compartments down its concentration gradient. The process of water movement across a selectively permeable membrane is called **osmosis**.

a. The balance of fluids into and out of the body

- The amount of water gained and lost each day varies, but, typically it is between 1600 mL and 2500 mL/day depending on the metabolic rate of the person, their body surface area and their weight.
- Other factors which can influence the amount of water taken in and lost, include diet, amount of exercise and external conditions (temperature and humidity can effect amount of water lost from the body).
- Below are the sources of water intake and loss from the body during a typical day:

| Water intake (2000 mL/day) | Water loss (2000 mL/day)                |
|----------------------------|-----------------------------------------|
| Food (560 mL)              | Breathed out from the lungs (240 mL)    |
| Metabolic water (160 mL)   | Skin through sweat (400 mL)             |
| Drink (1280 mL)            | Filtrate from kidneys (urine) (1200 mL) |
|                            | Alimentary canal (in faeces) (160 mL)   |

b. Excretion

- Excretion is the removal of poisonous waste products produced as a result of metabolic activity within cells.
- Excretory organs are structures which are used to remove poisonous waste products. These include:
  - o **Lungs:** Carbon dioxide and water are waste products of aerobic respiration. They are removed during gaseous exchange in the alveoli of the lungs.
  - o **Sweat glands:** The fluid (sweat) secreted onto the skin contains waste substances produced as a result of metabolism, including urea, salts and lactic acid.
  - o **Alimentary canal:** Bile is produced to emulsify fats for digestion by lipase. The pigments from this substance are the breakdown products of haemoglobin. Whilst faeces is made from undigested food and not as a result of metabolism within cells, the pigments are excretory products. In the main, faeces is egestion, **not** excretion.
  - o **Kidneys:** The bulk of the excretory material is removed via the kidneys, which are the main excretory organs. Urea is one of the main substances removed in this way. However, the majority of the excretory filtrate, or **urine**, is made up of water (91-95%).

| Composition of Urine                                                                                                             |
|----------------------------------------------------------------------------------------------------------------------------------|
| • Water (H <sub>2</sub> O): 95%                                                                                                  |
| • urea (H <sub>2</sub> NCONH <sub>2</sub> ): 9.3 g/l to 23.3 g/l                                                                 |
| • chloride (Cl <sup>-</sup> ): 1.87 g/l to 8.4 g/l                                                                               |
| • sodium (Na <sup>+</sup> ): 1.17 g/l to 4.39 g/l                                                                                |
| • potassium (K <sup>+</sup> ): 0.750 g/l to 2.61 g/l                                                                             |
| • creatinine (C <sub>4</sub> H <sub>7</sub> N <sub>3</sub> O): 0.670 g/l to 2.15 g/l                                             |
| • inorganic sulfur (S): 0.163 to 1.80 g/l                                                                                        |
| • Lesser amounts of other ions and compounds are present, including phosphorus, citric acid, ammonia, uric acid and many others. |

c. Osmoregulation by the kidneys

- The kidneys are able to regulate the amount of water lost and salts lost from the body, which neither the skin nor the alimentary canal are able to do.
- The kidneys are made up tiny tubules called **nephrons**.
- Nephrons are the site of filtration and re-absorption of the blood.
- The afferent arteriole leading to the glomerulus in each nephron is wider than the efferent arteriole leaving it. Consequently, the blood is under high pressure, so the small molecules that form the **glomerular filtrate** are squeezed into the glomerulus. From here, the fluid flows towards the ureter. Useful substances, such as water, some salts and all the glucose, are reabsorbed back into the blood.
- As much as 99% of the water filtered through the glomeruli of the nephrons is reabsorbed. Most of this occurs in the proximal convoluted tubules (PCT) and the loop of Henle by **osmosis**, so it is a passive process.
- Reabsorption in the distal convoluted tubules (DCT) and the collecting ducts requires energy from ATP, so it is **active reabsorption**. The degree of reabsorption possible is controlled by ADH, which increases the permeability of the DCT and collecting ducts to water. This is the active part of the process, which is also known as **facultative reabsorption**.
- The more ADH released, the greater the reabsorption and the more concentrated the urine is, which eventually passes down the ureter to the bladder. It is stored here until its release at an appropriate time.
- The action of ADH on the nephrons of the kidneys can be seen in the following feedback loop:

|           | <b>Lower water content in blood plasma</b>                                                                                                                                                      | <b>Higher water content in blood plasma</b>                                                                                                                                                    |
|-----------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Stimulus  | Water concentration of blood plasma decreases; osmotic pressure increases.                                                                                                                      | Water concentration of blood plasma increases; osmotic pressure decreases.                                                                                                                     |
| Receptors | Osmoreceptors in the hypothalamus detect increase in osmotic pressure.                                                                                                                          | Osmoreceptors in the hypothalamus detect decrease in osmotic pressure.                                                                                                                         |
| Modulator | Hypothalamus stimulates the posterior lobe of the pituitary gland, via nervous conduction, to release <b>more</b> ADH.                                                                          | Hypothalamus provides less stimulation of the posterior lobe of the pituitary gland, so <b>less</b> ADH is released. (Not 'no ADH released')                                                   |
| Effectors | The permeability of the walls of the DCT and collecting ducts is increased (through a greater number of water channels being opened).                                                           | The permeability of the walls of the DCT and collecting ducts is decreased (through less water channels being opened).                                                                         |
| Response  | More water is reabsorbed.<br>Less urine is produced and it is more concentrated.                                                                                                                | Less water is reabsorbed.<br>More urine is produced and it is less concentrated.                                                                                                               |
| Feedback  | There is an increase in water concentration in the blood. Therefore, the osmotic pressure is decreased.<br>The result is negative feedback as this eliminates or reduces the original stimulus. | There is a decrease in water concentration in the blood. Therefore, the osmotic pressure is increased.<br>The result is negative feedback as this eliminates or reduces the original stimulus. |

- **Aldosterone** also plays a role in regulating the balance of fluids. The release of this hormone results in the kidneys reabsorbing more sodium and excreting more potassium. Water is reabsorbed with sodium, so this, in effect, results in increased water reabsorption.
- Also, the increased water reabsorption increases the volume of blood plasma, which, in turn, increases the blood pressure.

**Extra information: How ADH increases water reabsorption by the DCT and collecting ducts**

- ADH binds to receptors in the plasma membrane of the cells lining the DCT and collecting ducts.
- This results in a series of enzyme-controlled reactions, which ends with the formation of an active phosphorylase enzyme.
- The phosphorylase enzyme causes vesicles, which are surrounded by membrane containing water-permeable channels, to move to the plasma membrane.
- The vesicles fuse with the plasma membrane allowing water to move freely through the membrane down its water concentration gradient, into concentrated tissue fluid and blood plasma in the medulla of the kidney.

d. Osmoregulation as a result of a behavioural response

- An increase in osmotic pressure in the extracellular fluid as a result of excess water loss stimulates the thirst response. The events can be seen in the table below.

| Regulating water intake behaviourally |                                                                                                                                                                                                             |
|---------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Stimulus                              | Water concentration of extracellular fluids decreases; osmotic pressure increases.                                                                                                                          |
| Receptors                             | Osmoreceptors in the thirst centre of the hypothalamus are stimulated.<br>Other stimuli, such as a dry mouth, are involved.                                                                                 |
| Modulator                             | The cerebrum detects the sensation of feeling thirsty, i.e. there is a conscious feeling to want to drink.                                                                                                  |
| Effectors                             | The person responds by having a drink.<br>Water drunk enters the bloodstream via the colon (large intestine); part of the alimentary canal.                                                                 |
| Response                              | Water concentration in the extracellular fluids increases; osmotic pressure decreases.<br>The water leaves the blood and the intercellular and intracellular fluids return to normal osmotic concentration. |
| Feedback                              | The result is negative feedback as the thirst centre is no longer stimulated, so the desire to drink ceases.                                                                                                |

e. Dehydration and water intoxication

- An excessive loss of water from the body accompanied by the loss of salts results in **dehydration**, i.e. more water is lost than is taken in.
- It often occurs during exercise when a lot of water is lost through sweat, or during illness, as a result of water loss from diarrhoea or vomiting.
- Most people can cope with mild dehydration, which is about a 3-4% loss of total body water. Greater than this usually results in fatigue and dizziness. Any loss of total body water greater than 10% can cause physical and mental deterioration. If this exceeds 15%, death is a likely outcome.
- Mild dehydration is usually resolved with oral rehydration.
- Care is needed when rehydrating, because it is possible to drink too much water, whilst not replacing the salts that have also been lost from the body. Salt and other electrolytes need to be replaced as well as rehydration. This condition of poisoning the body by drinking too much water is called **water intoxication**. It is rare.
- The first symptom of water intoxication is the sensation of light headedness. Headaches, vomiting and collapse may follow. Initial treatment involved restricting fluids. Then fluids with electrolytes to replenish those lost are taken.
- The most vulnerable people to dehydration are the elderly, because they have a reduced sensitivity to the thirst.
- Marathon runners are susceptible to water intoxication if they drink too much water during a run, as they lose a lot of salt and other electrolytes in sweat.

**Extra information**

- **Diabetes insipidus** is a rare condition, which mainly results from a deficiency of ADH.
- The main symptoms are excessive thirst and the production of large amounts of dilute urine, with reduction in fluid intake having no effect on the concentration of the urine.
- A second type of diabetes insipidus is caused by nephron or kidney dysfunction. This results in the kidneys not responding to ADH.

3. Blood glucose regulation

- Carbohydrates pass through the bloodstream in the form of glucose.
- All metabolizing cells require glucose in order to function, as it is the main respiratory substrate.
- The nervous system is especially sensitive to any reduction in blood glucose levels. A rise in blood glucose levels can be equally dangerous.
- The set point for blood glucose is 90 mg of glucose per 100 mL of blood.
- During a 24-hour period, the concentration of glucose in the blood fluctuates, because, we don't eat continuously, and, each meal contains varying amounts of glucose. Therefore, there can be periods of time when the bloodstream doesn't receive any glucose from the small intestines. However, cells need a constant supply of glucose to maintain metabolism.
- To this end, the liver plays a key role in maintaining a constant supply of glucose, by adding glucose to the blood in one of two ways:
  - o **Glycogenolysis**: the breakdown of glycogen to glucose.
  - o **Gluconeogenesis**: the conversion of other substrates, such as fats or amino acids to glucose.

a. How blood glucose is regulated

- As stated earlier, the organ which mainly regulates blood glucose concentrations is the pancreas.
- Regulation is controlled by the release of **insulin** from the  **$\beta$ -cells** of the islets of Langerhans in the pancreas, or **glucagon** from the  **$\alpha$ -cells** of the islets of Langerhans of the pancreas.
- In the table below is how these two hormones regulate blood glucose control:

|           | <b>Glucose levels too high (hyperglycaemia)</b>                                                                                                                                                                                                                                                                              | <b>Glucose levels too low (hypoglycaemia)</b>                                                                                                                                                                                     |
|-----------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Stimulus  | After a meal, there is an increase in the amount of glucose entering the blood from the small intestines.                                                                                                                                                                                                                    | When not eating, respiration still occurs, so the blood glucose levels start to fall below the set point.                                                                                                                         |
| Receptors | Detected by beta cells                                                                                                                                                                                                                                                                                                       | Detected by the alpha cells.                                                                                                                                                                                                      |
| Modulator | Beta cells of islet of Langerhans secrete insulin into the blood                                                                                                                                                                                                                                                             | Alpha cells of islets of Langerhans secrete glucagon into the blood.                                                                                                                                                              |
| Effectors | <ul style="list-style-type: none"> <li>• Glucose is taken up by cells (mainly of the liver and skeletal muscles).</li> <li>• Glucose is converted to glycogen (Process = <b>glycogenesis</b>)</li> <li>• Some glucose is converted to fat in adipose tissue</li> <li>• Protein synthesis increases in some cells.</li> </ul> | <ul style="list-style-type: none"> <li>• Stored glycogen in the liver is converted to glucose. (<b>Glycogenolysis</b>)</li> <li>• New glucose molecules are formed from fats and amino acids. (<b>Gluconeogenesis</b>)</li> </ul> |
| Response  | Lowers blood glucose levels.                                                                                                                                                                                                                                                                                                 | Increases blood glucose levels.                                                                                                                                                                                                   |
| Feedback  | The initial stimulus has been eliminated or reduced, so the $\beta$ -cells are no longer stimulated to release insulin. Feedback is negative.                                                                                                                                                                                | The initial stimulus has been eliminated or reduced, so the $\alpha$ -cells are no longer stimulated to release glucagon. Feedback is negative.                                                                                   |

- If the supply of glycogen from the liver becomes exhausted, glucose may be formed by other means.
- Once the low level of glucose in the blood is detected by the hypothalamus, it stimulates the anterior lobe of the pituitary gland to release **adrenocorticotrophic hormone (ACTH)**.
- This, in turn, causes the **adrenal cortex** to release **glucocorticoids (cortisol)**. This stimulates the conversion of glycogen into glucose in the liver, and protein breakdown in muscles, which allows for the conversion of amino acids into glucose.
- During stress, the **adrenal medulla** will release both adrenaline and noradrenaline, which results in the increase of glycogen breakdown. Also, adrenaline inhibits the effects of insulin and so less glucose is taken up from the blood by the cells.

b. Diabetes mellitus: A homeostatic disorder

- Hyperglycaemia is a result of blood glucose levels remaining too high.
- If the blood glucose levels remain too high, this person is suffering from **diabetes mellitus**, which is entirely different from diabetes insipidus.
- There are two types of diabetes mellitus;

|                       | <b>Type 1 or insulin-dependent diabetes</b>                                                                                                                                                                                                                                                                                                                                    | <b>Type 2 or non-insulin dependent diabetes</b>                                                                                                                                                                                                                                                                                                                           |
|-----------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Information           | Usually begins in childhood.Sometimes called <b>juvenile-onset diabetes</b> .                                                                                                                                                                                                                                                                                                  | Usually develops in later life, about 45 years old.<br>Sometimes called <b>adult-onset diabetes</b> . (Although more young people are being diagnosed with it).<br>More common in less active people or people with obesity.                                                                                                                                              |
| Problem               | Pancreas seems unable to secrete sufficient insulin.                                                                                                                                                                                                                                                                                                                           | Produces insulin, but the cells of the liver and muscles don't respond to it.                                                                                                                                                                                                                                                                                             |
| Symptoms              | <ul style="list-style-type: none"> <li>• Excessive thirst.</li> <li>• Frequent urination.</li> <li>• Weight loss.</li> <li>• Fatigue.</li> <li>• Visual disturbances, such as blurred vision.</li> <li>• Itching skin, particularly around the genitals.</li> <li>• Nausea and vomiting.</li> </ul>                                                                            | <ul style="list-style-type: none"> <li>• Excessive thirst.</li> <li>• Frequent or increased urination, especially at night.</li> <li>• Excessive hunger.</li> <li>• Fatigue.</li> <li>• Blurry vision.</li> <li>• Sores or cuts that won't heal.</li> </ul>                                                                                                               |
| Treatment/ Management | <ul style="list-style-type: none"> <li>• On most occasions the body will respond to insulin in the normal way, so regular injections of insulin or the use of an insulin pump are necessary.</li> <li>• Even with regular insulin injections, the long-term effects usually include kidney failure, heart attacks, strokes, amputations, blindness or nerve damage.</li> </ul> | <ul style="list-style-type: none"> <li>• Management of diet, including reduction of sugar and salt, in order to keep blood glucose levels in the normal range.</li> <li>• Regular exercise,</li> <li>• Maintaining a healthy weight.</li> <li>• Sometimes medication, such as Metformin, is needed, which improves the sensitivity of body tissues to insulin.</li> </ul> |

**Extra information: The development of synthetic insulin (Knowledge of synthetic hormones is in the Science for Human Endeavour part of the syllabus)**

- In 1921, Banting and Best isolated insulin from the pancreas of pigs and cows.
- They performed their experiments on dogs to measure sugar in their urine and blood.
- James Collip purified the insulin extract so that it could be used on humans.

- In 1922, Leonard Thompson became the first person to receive an insulin injection. It saved his life.
- In 1923, commercial production of insulin by Ili Lilly and Company and Nordisk Insulinlaboratorium began.
- In 1955, the first oral medication was made available, which stimulated the pancreas to release more insulin.
- In 1978, the first synthetic form of insulin was produced, whereby *E. coli* bacteria were used to produce insulin identical to human insulin. This reduced the side effects caused by non-human insulin. It was approved for commercial use in 1982.

#### 4. Regulation of gas concentrations

##### a. The breathing mechanism

- Before understanding how gases in the blood influence the rate of breathing, it is important to understand how breathing in (**inspiration**) and breathing out (**expiration**) is controlled.
- Within the medulla oblongata there is a region known as the **respiratory** (or **breathing**) centre.
- The ventral (or underside) of the respiratory centre is called the **inspiratory centre**; the rest controls breathing out and is called the **expiratory centre**.
- Also, control relies upon **chemoreceptors** in the **carotid and aortic bodies** of the blood system, which are sensitive to the slight changes in gas concentrations, mainly the concentration of carbon dioxide (more on this later).

Whilst inspiration and expiration are Unit 1 concepts, an understanding of the actions of the medulla oblongata in controlling normal breathing is in the syllabus. Therefore, it helps to revise the mechanism of breathing to understand how normal breathing is controlled and the factors affecting breathing.

##### i. Inspiration (Breathing in/Inhalation)

- Nervous impulses from the chemoreceptors stimulate the inspiratory centre of the medulla oblongata.
- This stimulates nerve impulses to pass along the **phrenic** and **intercostal (thoracic) nerves**.
- The phrenic nerve stimulates the muscles in the diaphragm to contract, lowering the diaphragm.
- The intercostal nerve stimulates the intercostal muscles to contract, causing the ribs to move up and out.
- This increases the volume of the thoracic cavity and decreases the pressure inside the lungs compared to atmospheric pressure, so air enters the lungs.
- The concentration of gases, particularly carbon dioxide concentration influences the rate that this occurs at, resulting in changes to the rate and depth of inspiration.

##### ii. Expiration (Breathing out/Exhalation)

- The expansion of the lungs causes **stretch receptors** in their walls to be stimulated.
- This results in nerve impulses travelling along the **vagus nerve**, which prevents further stimulation by the inspiratory centre, so the diaphragm and intercostal muscles relax, causing the diaphragm to return to its dome-shape and the ribs to move down and in.

- The thoracic cavity decreases in volume and the pressure inside the lungs is greater than atmospheric pressure, so air leaves the lungs, resulting in expiration.
- The stretch receptors are no longer stimulated and the expiratory centre is no longer stimulated, so the inspiratory centre can be stimulated again resulting in taking in the next breath.
- Breathing can be controlled to some extent by conscious thought from the forebrain, resulting in changes to the depth and rate of the breathing rate.
- However, whilst this allows us to control speech and to hold our breaths when necessary, we cannot stop breathing consciously forever. Eventually we will be forced to take a breath.

b. The effect of gas concentrations on breathing

- Both oxygen and carbon dioxide are carried in the blood. Both affect the rate of breathing, but it is **carbon dioxide concentration** that has the greatest affect.
- Oxygen is not very soluble in water, so 97% of it is carried by erythrocytes (red blood cells). It combines with haemoglobin to form oxyhaemoglobin. The other 3% is carried in the blood plasma.
- Carbon dioxide is carried in three ways:
  - o 8% is dissolved in the blood plasma.
  - o 22% combines with the haemoglobin in erythrocytes to form Carbaminohaemoglobin.
  - o 70% dissociates into hydrogen carbonate ( $\text{H}_2\text{CO}_3$ ) and then into hydrogen ions ( $\text{H}^+$ ) and hydrogen carbonate or bicarbonate ions ( $\text{HCO}^-$ ):

The equation for this dissociation is:



i. Oxygen concentration

- If the concentration of oxygen falls too low when all the other factors are held constant, the breathing rate increases.
- Changes on the breathing rate are very slight unless there is a significant drop in oxygen. Therefore, oxygen isn't the major gas influencing breathing rate.
- High altitude affects breathing rates because the change in the atmospheric pressure makes it more difficult to take the oxygen into our lungs, so we breathe harder and deeper.
- Changes in oxygen concentration are detected by the aortic bodies (three of which are found in the aorta and one of which is found in the right subclavian artery) and the carotid body (found in the external carotid artery). Only if there is a large change in the oxygen concentration will the aortic and carotid bodies be stimulated to send nerve impulses to the inspiratory centre of the medulla oblongata to speed up the breathing rate through contraction of the diaphragm and the intercostal muscles.

ii. Carbon dioxide concentration and hydrogen ion concentration

- Carbon dioxide is the major gas affecting the breathing rate.
- Unlike oxygen, a relatively small increase in carbon dioxide will stimulate the inspiratory centre of the medulla oblongata to increase breathing.
- An increase in carbon dioxide results in an increase in hydrogen ion concentrations and therefore a decrease in pH, i.e. the blood becomes more acidic.
- Both an increase in carbon dioxide and an increase in hydrogen ion concentration stimulate the peripheral and central chemoreceptors, which, in turn, stimulate the inspiratory centre resulting in an increased breathing rate.

- Whilst both high carbon dioxide concentration and a decrease in pH (higher acidity), both affect the peripheral and central chemoreceptors, the central chemoreceptors are more sensitive to changes in the concentration of carbon dioxide. The aortic and carotid bodies are more sensitive to a decrease in pH, as a result of an increase in hydrogen ion concentration.
- The central chemoreceptors are separate from, but communicate with the neurons of the respiratory centre, and they are responsible for 70-80% of the increase in the breathing rate. However, this takes a few minutes and so it is the peripheral chemoreceptors that are responsible for the initial increase in the breathing rate.

c. Other factors that impact breathing

i. Hyperventilation

- This is when a person suddenly breathes at an abnormally rapid rate.
- It can result in dizziness, light headedness and a sense of unsteadiness.
- It is common after strenuous aerobic exercise, but can also be caused by severe pain or emotional stress.
- When hyperventilating, more air is exhaled (breathed out) than is inhaled (breathed in).
- Consequently, there is less carbon dioxide in the blood, which results in a narrowing of the blood vessels to the brain. This, in turn, causes the light headedness and desire to take in more air.
- Whilst not the most effective or recommended method of preventing hyperventilation, some people breathe into a paper bag. The idea is that the person will breathe in more carbon dioxide, which will enter their blood, causing the blood vessels to widen, so allowing more blood to flow to the brain.
- It is not a good idea to hyperventilate before swimming under water. Whilst the reduction of carbon dioxide in the blood will result in a reduction in the breathing rate, meaning one can hold their breath for longer, the blood vessels narrow. This restricts the flow of oxygen to the brain. Consequently, there is an increased chance of going unconscious. On dry land, breathing returns to normal. Under water, breathing also return to normal, but instead of air, water is breathed in.

ii. Exercise

- During exercise, more energy is needed, so more glucose is broken down in the presence of oxygen and more carbon dioxide is produced (as well as water).
- Both the depth and rate of breathing are increased in order to take in more oxygen and remove more carbon dioxide.
- The same factors that influence breathing at rest are involved in increasing the breathing rate during exercise; namely, carbon dioxide concentration, pH change and, to a lesser extent, oxygen.



3. [10]  
 a. Complete the table below: (6)

| Hormone     | Endocrine Gland | Target Organ |
|-------------|-----------------|--------------|
| ADH         |                 |              |
|             | Adrenal medulla |              |
| Aldosterone |                 |              |

- b. Explain what aldosterone does. (1)

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- c. Name another hormone that is released from this endocrine gland and explain how it works. (3)

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4. Explain what the difference is between an exocrine and endocrine organ and give an example of an organ which is both exocrine and endocrine. [3]

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5. Metabolism is controlled hormonally. 1.3% of the population suffer from an increased basal metabolic rate as a result of a hormonal disorder. This increases to 4-5% in older women, although smoking can also result in an increase in this problem. One form of the disorder affects mostly younger women. [8]

- a. What is the overall name of this disorder? (1)

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- b. State **FOUR** symptoms of the condition. (4)

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- c. What treatment would you recommend for someone suffering from this disorder? (1)

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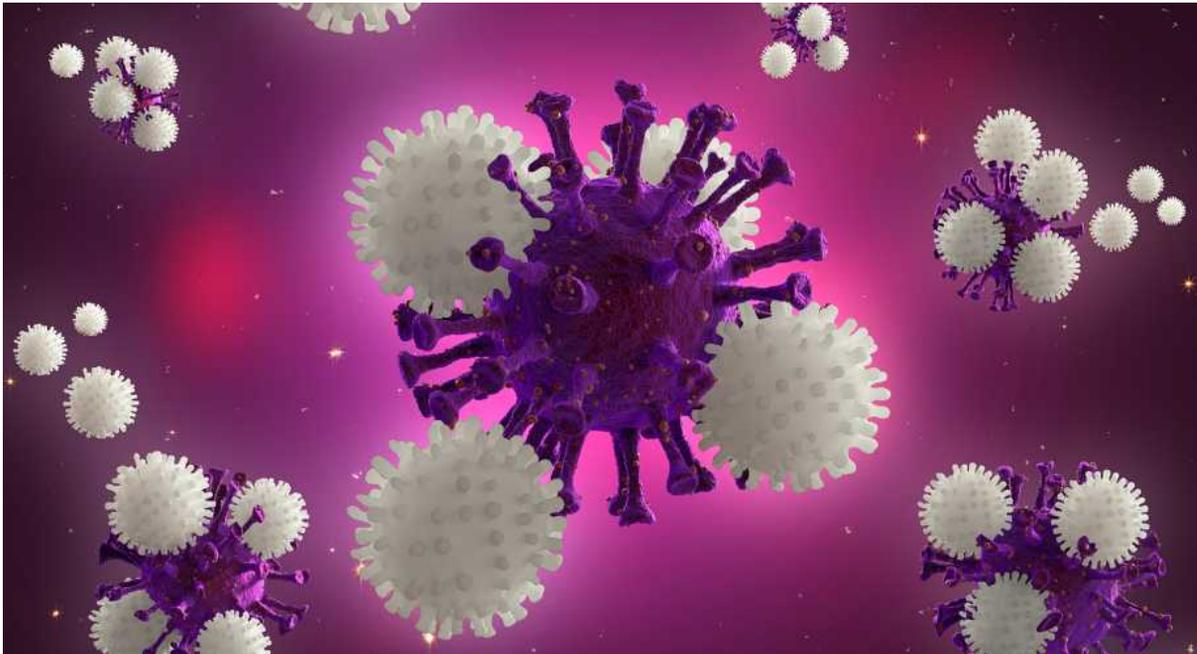






# RESPONSE TO INFECTION (UNIT 3)

# 4



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                                                      | Done | Revise | Teach others |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <ul style="list-style-type: none"> <li>I understand that infectious diseases caused by invasion of pathogens in the form of viruses and bacteria can be transmitted from one host to another.</li> </ul>                                                                                                                                                                 |      |        |              |
| <ul style="list-style-type: none"> <li>I know that transmission of pathogens occurs by various mechanisms, including through:                             <ul style="list-style-type: none"> <li>◦ direct and indirect contact</li> <li>◦ transfer of body fluids</li> <li>◦ disease-specific vectors</li> <li>◦ contaminated food and water</li> </ul> </li> </ul>      |      |        |              |
| <ul style="list-style-type: none"> <li>I understand that the body's external defence mechanisms against pathogens include features of the:                             <ul style="list-style-type: none"> <li>◦ Skin</li> <li>◦ digestive tract</li> <li>◦ urinogenital tract</li> <li>◦ respiratory system</li> <li>◦ the ear</li> <li>◦ the eye</li> </ul> </li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how pathogens that enter the body are targeted by nonspecific immune responses of inflammation and fever.</li> </ul>                                                                                                                                                                                                |      |        |              |
| <ul style="list-style-type: none"> <li>I know that antiviral and antibiotic drugs are used for treating infections and differ in their specificity to pathogens.</li> </ul>                                                                                                                                                                                              |      |        |              |

| Key teaching points                                                                                                                                                                                                                                                                | Done | Revise | Teach others |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <ul style="list-style-type: none"> <li>I understand that passive immunity can be acquired as antibodies gained through the placenta, or antibody serum injections; active immunity can be acquired through natural exposure to the pathogen, or the use of vaccines.</li> </ul>    |      |        |              |
| <ul style="list-style-type: none"> <li>I know that immunity is gained through the exposure to specific antigens by the production of antibodies by B lymphocytes and the provision of cell-mediated immunity by T lymphocytes; in both cases memory cells are produced.</li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can discuss the decision to participate in immunisation programs and that decisions can be influenced by the social, economic and cultural context in which it is considered. (SHE)</li> </ul>                                            |      |        |              |

### Helpful hints:

- Questions on disease lend themselves to extended responses very well. It could be broken down to:
  - Non-specific immunity.
  - Specific immunity.
  - Immunisation. This usually involves use of genetically modified vaccines.
  - Also, there are many ethical considerations with use or non-use of immunisation, which have been popular in the press and on social media.

### Key points:

- The body has three lines of defence.
  - External: **Non-specific**.
  - Internal: **Non-specific**.
  - Internal: **Specific**. This can be divided further into **humoral** and **cell-mediated** responses.
- Immunity is the ability of the body to resist or protect against infection caused by non-self (foreign) antigens. Immunity can be:
  - Natural** – received without intervention or **artificial** – received as a result of intervention
  - Passive** – antibodies are not produced by the infected person or **active** – antibodies are produced by the person infected in response to a specific, non-self antigen.

## INTRODUCTION

- The World Health Organisation struggles to define health simply. It is more than the absence of disease. “**Health** is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.”<sup>1</sup> (World Health Organisation, 2003)
- The human body has a number of mechanisms designed to prevent infection by invading organisms.
- Pathogens** are disease-causing organisms. Usually, they are very small and cannot be seen with the naked eye, i.e. a microscope is needed to see them. These are called **microorganisms**.
- Any disease that can be that is caused by a foreign (non-self) organism is said to be **communicable, transmissible** or **infectious**.
- The disease is said to be **contagious** if it is passed to another person by **direct contact** with an infected person.
- Vectors** are intermediate hosts of a pathogen, which act as an agent to pass on diseases. Examples include mosquitoes and fleas. *Plasmodium falciparum* is a protozoan parasite

1 World Health Organisation. (2003). *WHO Definition of Health*. Retrieved December 29, 2016, from WHO: <http://www.who.int/about/definition/en/print.html>

(causes harm to its host), which is known to cause malaria in humans. It is passed on by female *Anopheles* mosquitoes that bite humans in order to get a meal of blood. The infected bite contains the *P. falciparum*, which multiplies in the liver, causing severe damage to red blood cells. Eventually, this causes death.

- **Non-communicable diseases** can be caused by environmental, genetic or other factors which don't involve a specific pathogen. Often these "diseases" are known as **disorders** to distinguish them from communicable diseases.

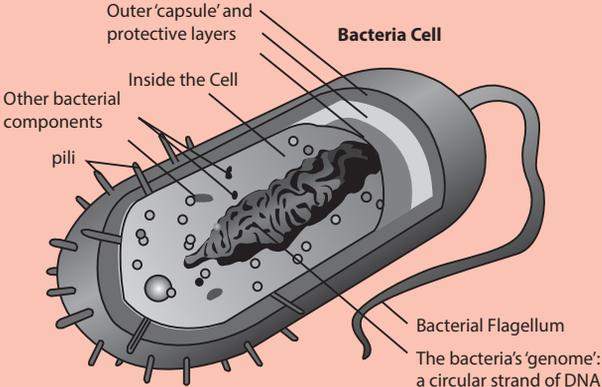
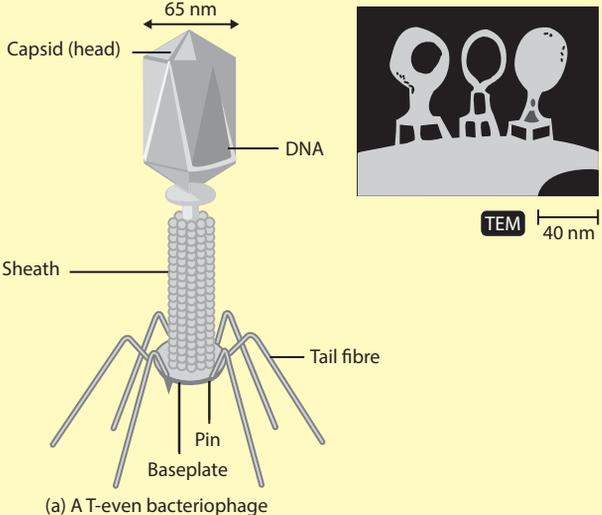
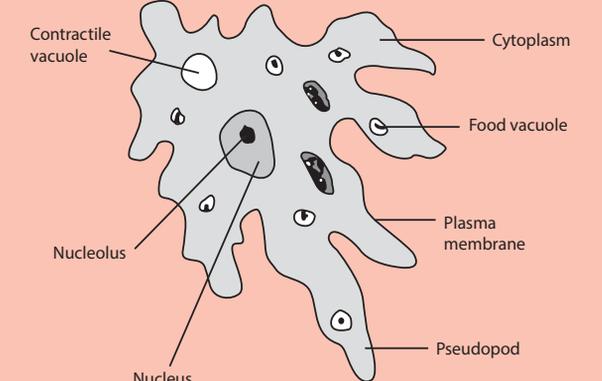
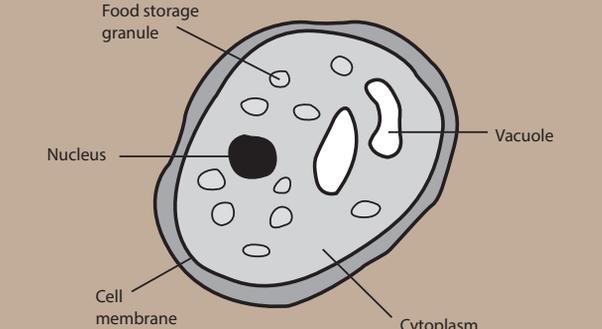
## TYPES OF PATHOGENS

There are 4 main types of pathogen; only bacteria and viruses are in the syllabus (see next page).

- Bacteria colonies (Singular: bacterium) are given different names dependent on their shape:
  - **Cocci**: Spherical bacteria
  - **Bacilli**: Rod-shaped (and have flagella for movement)
  - **Spirilla**: Have twisted cells.
  - **Vibrio**: "Tear-shaped" or "comma-shaped" cells.
- Viruses share most of the characteristics of living organisms, but due to their inability to reproduce independently, without the use of another organism as a host, they are mostly considered non-living. Viruses usually reproduce in the following way:
  - They attach themselves to a host cell and inject their DNA or RNA (only one or the other).
  - Inside the cell, replication of their genetic material occurs due to the presence of viral RNA polymerase.
  - Re-assembly of the virus occurs within the cell with the formation of new protein coats.
  - The new viruses are released by rupturing the cell, which usually causes the host cell to die. (This is not the case for the influenza virus, which keeps the host cell alive so that it can continue to produce new virus cells).
- **Bacteriophages** are viruses which attack bacterial cells. They have the nucleic acid DNA. Bacteria use enzymes which cut the DNA or RNA of viruses at specific sites. These are called **restriction enzymes** or **restriction endonucleases**.

### *Extra information*

- **Retroviruses**, probably the best known of which is Human Immunodeficiency Virus (HIV), contain RNA instead of DNA, but they are able to make a DNA complementary copy of their RNA once inside the host cell. They do this by using reverse transcriptase.
- The DNA form of the retrovirus genes is called the **provirus**. It is this that can be incorporated into the host's DNA. Here it can remain later for long periods of time, before it is expressed and new viral RNA are produced. Also, any division of the host cell results in the proviral DNA being replicated too. This is why people infected with HIV may display no symptoms for many years before they develop full-blown AIDS.
- Another problem with the proviral DNA is that it can impact on the host's DNA in the immediate vicinity to where it is located. This can result in a gene being activated ("switched on") causing the formation of malignant growths, i.e. **cancer**. Host cells which have acquired retroviruses in this way are called **oncogenes**.
- Retroviruses can cause diseases other than cancer, but these are mostly harmless. However, they remain in the host-cell DNA and so are passed on from one generation to the next via gametes. Such viruses are called **endogenous viruses** (See Unit 4 for more on this as a method for identifying common ancestors).

| Micro-organism           | Structure                                                                                                             | How it spreads disease?                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
|--------------------------|-----------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Bacteria                 |                                     | <p>Can be spread by person to person contact, in water droplets breathed in from the air, through infected food and through touching infected surfaces.</p> <p>Examples of pathogenic bacteria:</p> <ul style="list-style-type: none"> <li>• Bacterial meningitis</li> <li>• Cholera</li> <li>• Diphtheria.</li> <li>• Tuberculosis.</li> <li>• Typhoid.</li> </ul>                                                                                                                                                                                                                                                                                             |
| Virus                    |  <p>(a) A T-even bacteriophage</p> | <p>3 main ways:</p> <ol style="list-style-type: none"> <li>1. Person to person: Viruses that cannot survive for long outside of a host. Travel in body fluids. E.g. HIV</li> <li>2. Environment to person: Viruses that can survive outside of the body. E.g. Rhinovirus (the common cold) Can survive for about two days on some surfaces.</li> <li>3. Animal to person: The least common by far. <ul style="list-style-type: none"> <li>• Examples of viral pathogens include: <ul style="list-style-type: none"> <li>◦ Avian flu</li> <li>◦ Chickenpox</li> <li>◦ Colds.</li> <li>◦ Ross River Virus.</li> <li>◦ Rubella.</li> </ul> </li> </ul> </li> </ol> |
| <b>Extra Information</b> |                                                                                                                       |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| Protozoan                |                                   | <p>Passed on by ingesting infected water or through narrow nasal capillaries under high pressure. E.g. Amoebic dysentery.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Fungi                    |                                   | <p>Fungi release spores and most human fungi infections are as the result of opportunity. E.g. Low resistance to fight infection. Examples of pathogenic fungi include Candida and Athlete's foot.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                          |

## THE TRANSMISSION OF DISEASE

- Pathogens are passed on in a number of ways:

| Mode of transmission         | Description                                                                                                                                                                                                                                                                                                                                                                                              |
|------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Contact: Direct and indirect | <ul style="list-style-type: none"> <li>Touching an infected person and then putting your fingers in your mouth. (<b>Direct</b>).</li> <li>Touching an item that an infected person has touched and then putting your fingers in your mouth (before washing your hands). (<b>Indirect</b>), from using unwashed plates.</li> <li>Examples: skin infections like plantar warts, conjunctivitis.</li> </ul> |
| Transfer of body fluids      | <ul style="list-style-type: none"> <li>This could be the transfer of infected blood or fluids, like semen or substances produced by mucous membranes, e.g. saliva.</li> <li>Examples: HIV, glandular fever, hepatitis B and C.</li> </ul>                                                                                                                                                                |
| Infection by droplets        | <ul style="list-style-type: none"> <li>This is when pathogens are expelled in tiny droplets of moisture from an infected person. This is usually into the air through coughing, breathing, or sneezing, but droplets may settle on non-living items, like plates or utensils, which are then placed into food which is ingested.</li> <li>Examples: Measles, mumps, colds and influenza.</li> </ul>      |
| Disease-specific vectors     | <ul style="list-style-type: none"> <li>Some pathogens are transmitted from one organism to another by using another animal.</li> <li>Examples: Malaria, Lyme disease, Ross river virus.</li> </ul>                                                                                                                                                                                                       |
| Ingestion                    | <ul style="list-style-type: none"> <li>Food that is contaminated may not be cooked properly. Therefore, the pathogen has been given ideal conditions to grow and multiply.</li> <li>Also, some foods are left for too long and this gives time for bacteria in the air to settle in the food and grow.</li> <li>Examples: Dysentery, <i>Salmonella</i> and typhoid.</li> </ul>                           |

## DEFENCE AGAINST DISEASE

### a. Overview

- Defence against disease can be **non-specific**, working against all pathogens, or **specific**, which target one particular pathogen.
  - Non-specific barriers include skin, mucous membranes, tears, sebum, cilia.
  - Some lymphocytes and macrophages can be involved in non-specific defence.
- Phagocytes are involved in both non-specific and specific immunity. They engulf and digest micro-organisms using lysosomes.
- B cells and T cells offer specific immune response.

### b. Non-Specific Defences

#### i. External defences

- This is the body's first line of defence.
- The purpose of these barriers is to prevent pathogens from entering the bloodstream in the first place.
- Obviously, there are some naturally-occurring openings that are necessary for specific functions, which provide opportunities for pathogens to invade. These have defence mechanisms as well.

| Part of the body   | Explanation                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            |
|--------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Skin               | <ul style="list-style-type: none"> <li>It forms a waterproof mechanical barrier to pathogens, providing it isn't broken by cuts and/or abrasions.</li> <li>It has bacterial colonies on the surface which make it harder for invading pathogens to establish themselves.</li> <li><b>Sebum:</b> An oily secretion that contains is slightly acidic, making the pH of the skin between 4.5 and 6.2. This acidity makes the environment hostile to many pathogens.</li> <li><b>Sweat:</b> Contains salts and fatty acids that prevent many pathogens from being able to grow. Like the sebum,</li> </ul> |
| Digestive tract    | <ul style="list-style-type: none"> <li><b>Mucous membranes:</b> Secrete <b>mucus</b> onto inner lining of the digestive tract. This prevents entry of many micro-organisms, because it is sticky.</li> <li><b>Saliva:</b> In the mouth, can create a flushing and cleansing action. Also, they contain <b>lysozymes</b></li> <li><b>Hydrochloric acid</b> in the stomach: Creates a hostile environment for many micro-organisms, which are killed by the acids.</li> </ul>                                                                                                                            |
| Urinogenital tract | <ul style="list-style-type: none"> <li><b>Mucous membranes:</b> Secrete mucus to prevent entry of micro-organisms.</li> <li><b>Acid secretions</b> are released in the vagina, creating a hostile environment for micro-organisms.</li> <li><b>Flushing action:</b> In the <b>urethra</b> (tube leading from the bladder), this action prevents pathogens from building up and from travelling up towards the bladder and kidneys.</li> </ul>                                                                                                                                                          |
| Respiratory system | <ul style="list-style-type: none"> <li><b>Hairs</b> and <b>mucus</b> in the nasal cavity trap micro-organisms.</li> <li><b>Cilia</b> are tiny hairs in the trachea and bronchi that work together to trap microorganisms and dust particles and move them out of the respiratory tract with wave-like contractions. The mucus is either swallowed or coughed up.</li> </ul>                                                                                                                                                                                                                            |
| The ear            | <ul style="list-style-type: none"> <li><b>Hairs</b> in the ear prevent the entry of micro-organisms</li> <li><b>Cerumen</b> (Ear wax): Is slightly acidic and contains lysozymes (hydrolytic enzymes which can break down bacteria)</li> </ul>                                                                                                                                                                                                                                                                                                                                                         |
| The eye            | <ul style="list-style-type: none"> <li><b>Flushing action:</b> Tears prevent bacteria from growing. Also, they contain <b>lysozymes</b>, which break down micro-organisms.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                  |

ii. Protective reflexes (This is extra information in this context, but reflexes are in the syllabus)

- A reflex is an automatic, involuntary response to a stimulus. Whilst these are not external mechanisms of protection in the same way as those described above, they do prevent infection by carrying out actions aimed at preventing infection from developing in the body.

| Type of reflex | Explanation                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
|----------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Sneezing       | <ul style="list-style-type: none"> <li>Stimulus: Irritation of the walls of the nasal cavity.</li> <li>Forceful explosion of air from the lungs results in the release of mucus, foreign particles and gases out through the nose and mouth.</li> </ul>                                                                                                                                                                                                        |
| Coughing       | <ul style="list-style-type: none"> <li>Stimulus: Irritation in the lower respiratory tract.</li> <li>Air is forced from the lungs to try and release or remove the irritant. Coughing often releases mucus and foreign particles out of the trachea and into the throat and mouth.</li> </ul>                                                                                                                                                                  |
| Vomiting       | <ul style="list-style-type: none"> <li>Stimulus: Can be psychological, as a result of eating something that contains bacterial toxins, or due to overstretching the muscles of the stomach, e.g. due to over-eating.</li> <li>It is the involuntary, forceful expulsion of the contents of one's stomach, usually through the mouth, but sometimes through the nose. It is caused by the contraction of the abdomen and diaphragm, not the stomach.</li> </ul> |
| Diarrhoea      | <ul style="list-style-type: none"> <li>Stimulation: Irritation of the walls of the small intestine and colon (large intestine).</li> <li>Water is usually absorbed in the colon, but the walls are inflamed and contract more readily, allowing less water to be absorbed. This is caused by the presence of foreign bacteria, viruses or protozoan. The faeces is more watery, leading to faster expulsion from the anus.</li> </ul>                          |

iii. Types of white blood cells (leucocytes) and internal non-specific defences

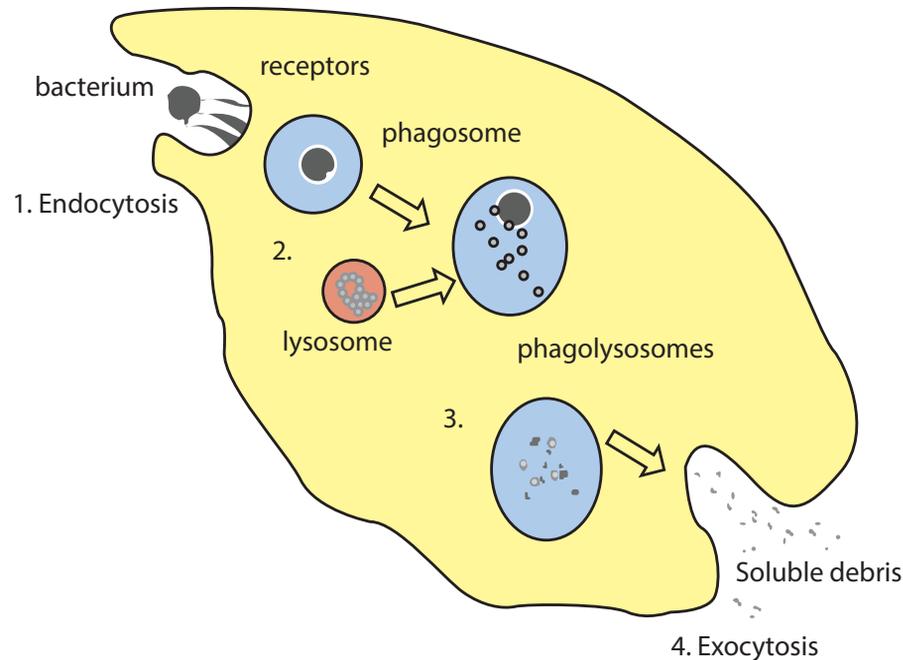
- There are three main types of white blood cells, all of which are involved in the defence against disease; granulocytes, monocytes and lymphocytes. The focus of this course is on the sub-class of monocytes, macrophages, and the two types of lymphocytes; B-cells and T-cells; these cells are emboldened in the table, as are processes which are covered later in the notes.

| Type of White Blood Cell                                       | Sub-class                                                                                                                                                                         | Overview of function                                                                                                                                                                              |
|----------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Granulocytes: Phagocytes that engulf and ingest foreign cells. | • Neutrophils                                                                                                                                                                     | Most common and ingest up to 5 and 20 bacteria.                                                                                                                                                   |
|                                                                | • Eosinophils.                                                                                                                                                                    | Involved in allergic reactions. Can attack multicellular parasites.                                                                                                                               |
|                                                                | • Basophils.                                                                                                                                                                      | Involved in allergic reactions and can release <b>histamines</b> , to trigger <b>inflammations</b> , and <b>heparin</b> , to prevent blood clotting.                                              |
| Monocytes                                                      | • Dendritic cells                                                                                                                                                                 | Antigen-presenting cells.                                                                                                                                                                         |
|                                                                | • <b>Macrophages</b> – Develop from some leucocytes. Some wander through the blood looking for pathogens; others are fixed in one place dealing with pathogens that come to them. | <b>Phagocytic cells which are larger and survive longer than neutrophils.</b>                                                                                                                     |
| Lymphocytes                                                    | • <b>B cells</b>                                                                                                                                                                  | Involved in humoral/antibody mediated specific immune response, providing resistance to viruses, bacteria and bacterial toxins before these micro-organisms or substances enter the body's cells. |
|                                                                | • <b>T cells</b>                                                                                                                                                                  | Involved in cell-mediated immunity, providing defence once the bacteria or viruses have invaded the body's cells.                                                                                 |

**How phagocytosis occurs**

1. The phagocyte is attracted to the foreign antigen/micro-organism (e.g. chemotaxis or by opsinization). In this example, the antigen is on the surface of a bacterium. Ingestion by the phagocyte/monocyte/macrophage/neutrophil, or endocytosis occurs.
2. A vacuole forms inside the phagocytic cell. A lysosome binds to the vacuole and releases digestive enzymes. When the lysosome is bound to the phagosome it is called a phagolysosome.
3. The digestive enzymes break down the microbe, leaving only soluble debris, which can be used by the body.

4. This leaves the phagocyte by exocytosis.



#### iv. The inflammatory response

- **Inflammation** is a response that occurs when tissue is damaged. This usually occurs when the skin has been breached in some way, e.g. piercing the skin with a pin. The damaged part of the skin becomes **red, swollen** and **hot**, and it is often accompanied by **pain**.
- Inflammation is aimed at reducing the spread of infection, destroying any microorganisms and reducing the risk of further infection by reducing the opportunity for more pathogens to enter the wound.
- During the inflammatory process, cell debris is removed and repair begins on the damaged tissue.
- Mechanical damage results in localised, chemical change whereby **mast cells** stimulate the production of **histamine, heparin** and other substances into the tissue fluid.
  - o **Histamine** increases blood flow to the site of injury and causes the capillaries in the area to become more permeable. With more blood flow, there is an increase in metabolic heat and the area becomes redder. Also, with more fluid being filtered from the blood, the site becomes swollen.
  - o **Heparin** prevents the blood clotting in this localised area. This allows the flow of blood to the injury to allow phagocytes to be attracted to the chemicals released by the mast cells. Blood clots do form around the damaged area, slowing down blood flow so that any pathogens present spread less quickly.
  - o **Phagocytes** are attracted to the site of injury by the chemicals released. Macrophages and leucocytes carry out phagocytosis.
- Often pain receptors are impinged on during this process, so the person feels pain.
- The phagocytic cells filled with the pathogens and debris begin to die, and, together with tissue fluid, form a yellow liquid called **pus**.
- Over time, new cells are formed by mitosis, and repair of the damaged tissue takes place.

#### v. Fever

- This is a homeostatic mechanism whereby the body's core temperature is reset at a higher temperature, due to an infection. This increase in temperature is called a **fever**.

- Due to the higher set point, the body feels cold and responds by attempting to increase the body temperature.
- This results in processes that relate to overcooling. Shivering generates metabolic heat and vasoconstriction of the blood vessels in the skin reduces heat loss, causing the body temperature to rise.
- If this continues, it will become very dangerous as biochemical processes in the body will break down. Death will result if the body temperature reaches 44.4-45.5 °C. To counteract this, medicine, such as paracetamol, is taken to lower the body temperature artificially.
- The benefits of the higher temperature are that it may disrupt the growth of some micro-organisms, and also it may result in faster biochemical reactions (up to a point), leading to quicker repair.
- Once the fever breaks, at the point called **crisis**, the body temperature is reset to normal, i.e. 36.8 °C. Consequently, the body feels too hot and mechanisms to cool the body down occur, i.e. vasodilation of the blood vessels in the skin and greater sweat production, which increase heat loss. The person tends to suffer from hot flushes.

#### *A word on pyrogens*

- Pyrogen literally means 'heat-' (pyro-) 'generator' (-gen).
- A pyrogen is a substance (infectious organisms or their product toxins or cytokines) that provokes fever.
- Exogenous pyrogens are substances which originate outside the body and are capable of inducing interleukins, e.g. interleukin-1, which are chemicals that help regulate immune responses.
- Endogenous pyrogens are substances, which originate inside the body and which are capable of inducing fever by acting on the thermoregulatory centre of the hypothalamus

#### *Extra reading*

##### The lymphatic system

- The lymph system is made up widely of distributed lymph capillaries that are found in all the tissues of the body. These tissues merge to form lymph vessels that possess valves and whose structure is similar to that of veins.
- The lymph is the fluid within these vessels, which is transported away from the tissues in one direction only. It transports antigen-presenting cells (APCs), such as dendritic cells, to the lymph nodes, which stimulates an immune response.
- A series of lymph nodes appear along the lymph vessels. They contain a population of phagocytic cells, which remove micro-organisms from the lymph.
- Lymph nodes are the major sites of lymphocyte production and, during infection, they frequently swell.
- One negative aspect of the lymph system is that it can over-swell and act as a home for bacterial infections, as well as spreading bacteria or cancerous cells.

##### Hygiene

- Most hygiene is common sense. The purpose of hygiene is to reduce the risk of infection by pathogens.
- Good practices include:
  - ✓ Washing your hands before and after preparing food, after using the bathroom, before and after providing first aid, after coughing and sneezing.
  - ✓ Covering your mouth when you cough or sneeze.

- ✓ Wearing gloves when handling blood or other bodily fluids.
- ✓ Wiping down surfaces with antiseptic or antibacterial sprays where food is to be prepared and after food or contaminated material has been used on the surface.
- ✓ Using tongs or tweezers to remove infected materials, such as used condoms or discarded syringes (as well as wearing protective gloves).
- ✓ Wearing masks to prevent coughing and sneezing over food or when carrying out medical procedures.
- ✓ Wearing hair nets when preparing food.

### c. The Immune Response

- Once the first line of defence is breached and the second line of defence is activated, **antigen presentation** occurs in the lymph system, which results in a specific immune response.
- Macrophages are involved in both non-specific and specific immunity. T-lymphocytes and B-lymphocytes are involved in the specific immune response. (See table of white blood cells earlier in this chapter).
- An **antigen** is any substance capable of producing a specific immune response. Antigens are substances on the surface of cell membranes. So, often they are proteins, but they can be carbohydrates or lipids.
- Antigens are found on the surface of all cells, including those produced by the individual. Those belonging to the individual are **self-** or **non-foreign antigens**. The body is programmed to recognise these and not attack them. Those not belonging to the body, e.g. on the surface of pathogens, are called **non-self** or **foreign antigens**.
- An autoimmune disorder is caused by the body not recognising self-antigens and attacking its own tissues.
- There are two types of specific immune response:
  1. **Humoral (cell-mediated) response**
    - o B cells are stimulated by a foreign substance (**antigen** (capable of producing an immune response)) which reaches **lymphoid tissue**. This is called **antigen presentation**.
    - o Sensitised B (lymphocyte) cells are produced and mature in bone marrow.
    - o B cells are stimulated and rapidly divide by mitosis to produce plasma cells and some memory cells.
    - o Plasma cells produce **antibodies** (specialised proteins produced in response to a non-self antigen) which are released into the bloodstream.
    - o Antibodies inactivate or destroy non-self antigens by:
      - Binding to viral binding sites or to bacterial antigens.
      - **Agglutination**: clumping non-self particles together.
      - Reacting with soluble antigens and making them insoluble.
      - Coat bacteria enhancing phagocytosis. (1<sup>st</sup> three points also enhance phagocytosis)
      - Inhibiting reactions with other cells or compounds by breakdown of non-self cell.
    - o This is a **primary response**. It takes time to develop the antibodies and so the individual usually suffers symptoms of the disease caused by the pathogen.
    - o Memory cells result in a much faster response should the same non-self antigen invade the body again.
  2. **Cell mediated response**
    - o This provides resistance to the intra-cellular phase of bacterial and viral infections.
    - o Foreign antigens reach lymphoid tissue.

- o T cells are sensitised and undergo rapid cell division. Most of the cells produced are **killer T cells**, which destroy the antigen. Other T cells become **helper T cells**, which sensitise lymphocytes, intensify response and promote phagocytosis, and **suppressor T cells**, which inhibit B and T cells to slow down the immune response once the infection has been dealt with effectively.
- o Some T cells form memory cells which initiate a faster **secondary response** should the same antigen re-enter the body.
- Secondary responses are always faster than primary responses, because there are a few memory cells moving around in the blood plasma. When the same non-self antigen attacks the body again, antibodies are produced rapidly and remain in the plasma for a lot longer. Consequently, the symptoms of the disease are not normally experienced.

**Types of Immunity**

- Immunity is the resistance to infection by invading micro-organism.
- Immunity can be **natural**, i.e. there is no human intervention, or **artificial**, i.e. as a result of being given an antibody or antigen.
- **Passive** immunity is when the body receives antibodies from another source. **Active** immunity is when the body produces its own antibodies in response to a non-self antigen.

|                | Natural                                                                                                                                                                                                                                                                                                 | Artificial                                                                                                                                                                                                                                                                                                                                         |
|----------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Passive</b> | <ul style="list-style-type: none"> <li>• Occurs without human intervention.</li> <li>• Body gets antibodies produced by someone else.</li> <li>• Not long lasting.</li> <li>• Example: antibodies enter the bloodstream across the placenta or in breast milk.</li> </ul>                               | <ul style="list-style-type: none"> <li>• Occurs when someone is given antibodies.</li> <li>• Body gets antibodies produced by someone else.</li> <li>• Not long lasting.</li> <li>• Example: influenza antibodies injected into bloodstream.</li> </ul>                                                                                            |
| <b>Active</b>  | <ul style="list-style-type: none"> <li>• Occurs without human intervention.</li> <li>• Body makes its own antibodies in response to a non-self antigen.</li> <li>• Long lasting immunity because of memory cells.</li> <li>• Example: Contracting chickenpox and producing antibodies to it.</li> </ul> | <ul style="list-style-type: none"> <li>• Occurs when someone is given antigens.</li> <li>• Body makes its own antibodies in response to a non-self antigen.</li> <li>• Long lasting immunity because of memory cells.</li> <li>• Example: Antigens given by vaccination, e.g. living attenuated MMR antigens injected into bloodstream.</li> </ul> |

- A **vaccine** is an antigen preparation used in artificial immunisation.

| Type of vaccine                   | Explanation and examples                                                                                                                                                                                                |
|-----------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Living attenuated micro-organisms | <ul style="list-style-type: none"> <li>• Reduced virulence (less ability to produce the disease).</li> <li>• Can be produced by recombinant DNA technology.</li> <li>• Examples: MMR, rabies, poliomyelitis.</li> </ul> |
| Dead micro-organisms              | <ul style="list-style-type: none"> <li>• Microbe killed before being injected.</li> <li>• Not as long-lasting but can result in an immune response.</li> <li>• Examples: cholera, bubonic plague, typhoid.</li> </ul>   |
| Toxoids                           | <ul style="list-style-type: none"> <li>• The toxins produced by bacteria can be inactivated so they don't make the person ill.</li> <li>• Examples: diphtheria, tetanus.</li> </ul>                                     |
| Sub-unit                          | <ul style="list-style-type: none"> <li>• A fragment of the micro-organism is used to provoke an immune response.</li> <li>• Examples: human papilloma virus, hepatitis B.</li> </ul>                                    |

- Vaccines are usually delivered by injection, but may be delivered by sugary syrup or other methods. Research into nasal sprays is being conducted, as are skin patches and food supplements.
- **Herd immunity**: A high proportion of the population are immunised so those who are not immune are protected. For example: influenza immunity in winter.
- Good herd immunity depends on all people getting on board with the program and not becoming complacent. Otherwise herd immunity is less effective.

## USE OF ANTIBIOTICS AND ANTIVIRAL DRUGS

### a. Antibiotics

- **Antibiotics** are drugs that are used to fight infections caused by bacteria.
- Some are naturally occurring, but most are now manufactured synthetically.
- If they are effective on only a specific or small range of bacteria, they are known as **narrow spectrum antibiotics**. Others are effective in inhibiting the growth of a number of different bacteria. These are called **broad spectrum antibiotics**.
- Penicillin was the first antibiotic. It was discovered by Alexander Fleming in 1928, and derived from a fungal mould.
- In 1938, Howard Florey and his team, which included Ernst Chain and Norman Heatley, began their work on producing a stable form of penicillin, so that it could be mass produced.
- In the 1940s, Pfizer successfully mass produced pharmaceutical-grade penicillin.
- Also in the 1940s, other micro-organisms that produce antibiotic substances were discovered, notably **streptomycin**, in 1943. The **actinomycetes** are bacteria that live in the soil and produce branching filaments which can disrupt protein synthesis in the cells of the target bacteria. Many of the existing antibiotics are derived from actinomycetes.
- There are two types of antibiotics:
  - **Bactericidal antibiotics**, which kill bacteria directly by changing the structure of the cell wall or cell membrane, or by disrupting the action of essential enzymes.
  - **Bacteriostatic antibiotics**, which prevent bacterial cells from growing, usually by preventing protein synthesis.
- Overuse of antibiotics can lead to problems, because certain bacteria have developed resistance to them. Some people don't finish the course of antibiotics they are on, so some bacterial cells mutate and the antibiotic becomes ineffective. Also, some doctors prescribe antibiotics for preventing rather than treating bacterial infections. This has led to the development of so-called "superbugs", which are either resistant to several antibiotics (**multiple drug resistance**) or to all known drugs (**total drug resistance**). Consequently, the search is on to manufacture antibiotics to which certain bacteria have no resistance.

### b. Antivirals

- Antibiotics have no effect on viruses. Again, this is problematic when people ask for antibiotics for viral infections.
- **Antivirals**, as their name suggests, are used in the treatment of infections caused by viruses.
- Antivirals work by inhibiting the development of the virus.
- Most antivirals are targeted at HIV, hepatitis B and C, and influenza A and B.
- Viral infections like colds, chickenpox and measles have no treatment and it is recommended that paracetamol are taken and drinking hot drinks. These are more to soothe than to cure.
- Now that scientists have been able to understand the genetic sequence of viruses, it is likely that more antivirals will be available in the future.

## RISK OF VACCINES AND ETHICAL CONCERNS

- Some vaccines are considered to have possible side effects which may be dangerous. A poor example of this is MMR increasing the risk of autism in children. There is no evidence to support this, but it has resulted in some people insisting on separate measles, mumps and Rubella injections. Of course, the risk of death from these diseases is far higher. Misinformation can result in some poor decisions. It is important to not simply take on board popular opinion, but to check the actual evidence.
- Ethical concerns exist over the preparation of some vaccines and the testing of vaccines.

Before clinical trials are conducted, most vaccines are tested on animals. This can lead to moral dilemmas.

- Also, viruses can only reproduce in living cells, so viral vaccines require host tissues. For example, influenza virus is cultured in chicken embryos. This results in animal ethical concerns.
- Testing of vaccines causes some concerns, because, whilst most vaccines are produced in developed countries, they are most likely to be used in developing countries. Concerns centre around whether the test groups in these regions are fully aware of the risks involved; i.e. they may be exploited.
- Proponents of animal testing take the view that several breakthroughs in medicine have come about as a result of testing on animals first
- Parents need to assess the risks of vaccination, where side effects or an allergic reaction may result in permanent damage or even death. Most parents consider the benefits outweigh the potential risks involved. This results in herd immunity in a number of cases and so reduces the risk of passing on the disease from an infected person.
- Australia was one of the first countries to roll out a national cervical cancer immunisation using Gardasil®. Gardasil® protects against certain strains of human papilloma virus (HPV) and is most effective when used before girls become sexually active, around 11 or 12. It does have some side effects in some cases, with girls experiencing fever, headaches, nausea and muscle or joint pain. Most health professionals and parents believe this age is too young to be discussing such matters. However, taking Gardasil® could prevent serious illness later in life. This is an example of being fully informed of the issues before making a decision.
- In Australia, immunisation programmes are mostly free, but this is not the case in all countries. Economic reasons may prevent some individuals being immunised against a particular pathogen.
- Poor education in some developing countries can result in some individuals not getting immunised or parents not immunising their children.
- Misinformation, as mentioned earlier, can result in people avoiding immunisation. This is particularly the case with the increase in social media. It is important to check the reliability of sources and not simply assume that what is presented is correct.
- The increase in popularity in alternative medicines has resulted in some people believing that conventional medicine, including vaccines, is associated with more risks than benefits. This is concerning when scientific evidence points to the benefits outweighing the potential risks.
- In some cultures, alternative medicine is given greater credence than conventional medicine, and parents are happy to rely on the advice of traditional healers, even when there is no scientific evidence to support the effectiveness of this alternative medicine.





# QUESTIONS

45 marks

1. What is meant by the following terms? [4]

a. Pathogen.

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b. Bacteriophage.

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c. Cilia.

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d. Lysozyme.

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2. Explain the difference between non-specific and specific defence against disease. [2]

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3. Describe THREE protective reflexes. [3]

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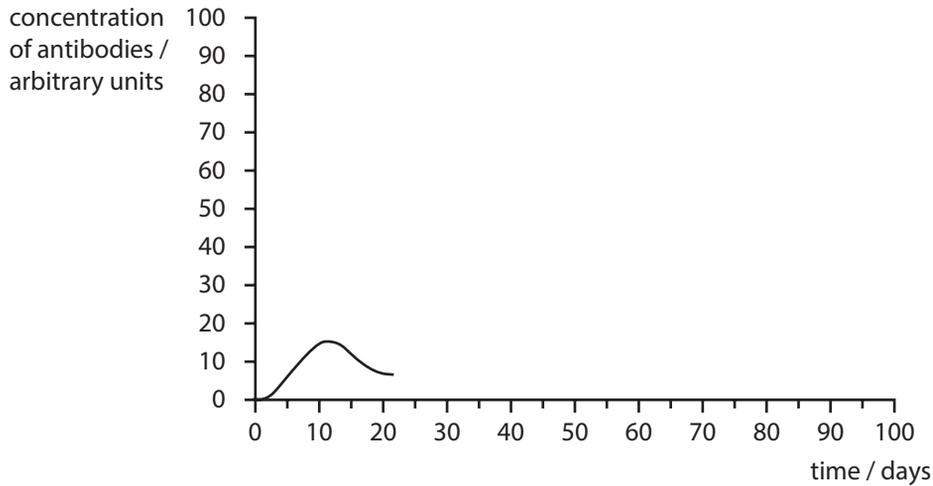
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6. Look at the graph below: [6]



a. Explain why there is a delay by a couple of days between the infection, which began on day 0, and the first appearance of antibodies in the blood. (2)

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b. On day 25, the person was exposed to a secondary infection by the **same** pathogen. Complete the graph to show the expected concentration of antibodies. (2)

c. Explain why this person experienced symptoms on the first exposure, but not the second exposure to this infection. (2)

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# MUTATIONS AND GENE POOLS (UNIT 4)

# 5



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                           | Done | Revise | Teach others |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Mutations (Unit 4)</b>                                                                                                                                                                                                                     |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain that mutations in genes and chromosomes can result from errors in DNA replication, cell division or from damage caused by mutagens.</li> </ul>                                           |      |        |              |
| <ul style="list-style-type: none"> <li>I know that different genotypes produce a variety of phenotypes, which are acted on differently by factors in the environment, producing different rates of survival.</li> </ul>                       |      |        |              |
| <ul style="list-style-type: none"> <li>I understand that mutations are the ultimate source of variation introducing new alleles into a population.</li> </ul>                                                                                 |      |        |              |
| <ul style="list-style-type: none"> <li>I can show that new alleles may be favourable or unfavourable to survival.</li> </ul>                                                                                                                  |      |        |              |
| <b>Gene pools (Unit 4)</b>                                                                                                                                                                                                                    |      |        |              |
| <ul style="list-style-type: none"> <li>I can demonstrate that populations can be represented as gene pools that reflect the frequency of alleles of a particular gene.</li> </ul>                                                             |      |        |              |
| <ul style="list-style-type: none"> <li>I can show that gene pools can be used to compare populations at different times or locations.</li> </ul>                                                                                              |      |        |              |
| <ul style="list-style-type: none"> <li>I know that gene pools are dynamic, with changes in allele frequency caused by:                             <ul style="list-style-type: none"> <li>mutations</li> </ul> </li> </ul>                    |      |        |              |
| <ul style="list-style-type: none"> <li> <ul style="list-style-type: none"> <li>differing selection pressures</li> </ul> </li> </ul>                                                                                                           |      |        |              |
| <ul style="list-style-type: none"> <li> <ul style="list-style-type: none"> <li>random genetic drift, including the founder effect</li> </ul> </li> </ul>                                                                                      |      |        |              |
| <ul style="list-style-type: none"> <li> <ul style="list-style-type: none"> <li>changes in gene flow between adjoining groups</li> </ul> </li> </ul>                                                                                           |      |        |              |
| <ul style="list-style-type: none"> <li>I understand that the incidence of genetic diseases in particular populations illustrates the effects of different factors on the dynamics of gene pools, for example, sickle-cell anaemia.</li> </ul> |      |        |              |

**Helpful hints:**

- Questions about mutations are often linked with other mechanisms of evolution.
- Often questions highlighting differences between mutations are given, e.g. the difference between a chromosomal and a point mutation, or a germline and a somatic mutation.

**Key points:**

- Mutations result in differences in between individuals within a species. Some of these differences may provide a selective advantage, whilst most result in disadvantages.
- Point mutations are likely to result in a change in allele frequency over time.
- Chromosomal mutations may allow an individual to survive, but they are likely to have some disabilities as a result of the extra information caused by non-disjunction in meiosis.

**INTRODUCTION**

- Meiosis is a process which results in new cells being formed from existing cells. In humans, one **diploid** cell (a cell with each chromosome paired with its corresponding chromosome) forms a tetrad (four) of **haploid** cells. Therefore, these cells have half the number of chromosomes of the parent cells.
- During sexual reproduction, the male gamete (sperm) fuses with the female gamete (ovum) to form a diploid cell. This cell undergoes mitotic division several times to eventually form a new individual.
- The new individual receives information contained on the chromosomes from both parents. Some of this information is expressed, whilst other pieces of information are present, but not expressed.
- Those pieces of information that are always expressed in the **phenotype** are called **dominant**. Those that are only expressed if the pieces of information at a specific point (or **locus**) on the homologous chromosomes are the same are called **recessive**.
- A **gene** is a section of DNA that contains the information that codes for a particular characteristic.
- Changes may occur within that section of DNA, which result in the formation **over time** of alternative forms of the gene. The changes that occur at a single point within a section of DNA are called **point mutations** or **gene mutations**. The alternative forms of the gene that arise are called **alleles**.
- Whilst DNA replication is highly reliable, it is possible for an incorrect nitrogenous base pair to be inserted, to be substituted, or to be deleted during the process. Then, when protein synthesis occurs, this change may result in the insertion of a different amino acid, altering the shape of the overall protein. In the case of deletion, there is a frame shift in the resulting mRNA codon. Therefore, the length of the resulting amino acid chain could be shorter or longer, because the STOP codon is no longer in the same place. This usually results in self-abortion. Usually gene mutations are recessive, so they are masked by the dominant gene and aren't seen in the phenotype. These mutations are called **lethal recessives** (See below).
- Sometimes, during anaphase I, the chromosomes (or part of a chromosome) within a pair fail to separate, or in anaphase II, the sister chromatids don't separate. This results in half the ova containing an extra chromosome and the other half containing one less chromosome. This means that the resulting zygote formed when the ovum and sperm fuse wither has too much information (all or part of an extra chromosome) or too little information (part or none of a chromosome that should be present). This is a **chromosomal mutation**. Usually, the zygote will self-abort, but some individuals with an extra chromosome survive. An example is **trisomy 21** or Down's syndrome, where the individual has an extra chromosome 21.

## GENE POOLS

- A **species** is made up individuals that are able to interbreed to produce **fertile** offspring. However, whilst humans share many characteristics, slight differences occur, because of variation caused by the processes described above.
- A **population** is a group of individuals of the same species living within a particular area at the same time.
- For certain characteristics there can be two or more alleles. Whilst each individual can only receive two alleles for each characteristic expressed, there may be a number of possible alleles that could be received.
- When studying a particular characteristic within a population, geneticists recognise that, in theory, any one individual could breed with any other, of the opposite gender. In other words, the genes are freely interchangeable. As such, the total of all the alleles of all the genes within the population is called the **gene pool**.
- Within a specific gene pool the number of times any one allele for a particular characteristic occurs is referred to as the **allele frequency**.
- Allele frequencies for particular characteristics can vary between populations. For example, a particular point mutation may occur in an isolated population more often than in the general population. Due to the isolation of the population, the chances of this allele being passed on are higher than it would be in the general population, where the allele has a low frequency.
- When small populations are studied within a larger general population, it is possible to see differences in the allele frequency for particular traits. This is as a result of **random genetic drift**. Also, if a small group of individuals were isolated from the main population, they may contain differences in allele frequency for a particular trait compared to the population out of which they came. This is known as the **founder effect**. (More on random genetic drift and the founder effect in chapter 7).

## CAUSES OF MUTATIONS

- Mutations can occur without any reason, but certain agents are capable of increasing the chances of mutations occurring. These agents are called **mutagenic agents** or **mutagens**.
- Examples of mutagens include exposure to ionising radiation, e.g. too much exposure to X-rays, or too much exposure to certain chemicals, e.g. benzene, nitrous acid, mustard gas, or formaldehyde.
- Cigarettes contain over 300 cancer-causing chemicals, many of which are mutagens.
- Mutations can affect our body cells (somatic cells) or our reproductive cells.
- Mutations that affect our body cells are called **somatic mutations**. They affect the individual, e.g. they may cause cancer, but they cannot be passed on to the offspring, because they don't affect the reproductive cells.
- Mutations that affect the reproductive cells are called **germline** or **germinal mutations**. They don't usually affect the individual, but they are passed on in the gametes. Often, the zygote fails to develop, but diseases, such as **phenylketonuria (PKU)** can develop.
- Whilst many mutations are harmful, some have no obvious affect (benign) and still others are beneficial and may give a selective advantage if the environment is suitable.

## LETHAL RECESSIVES

- These point mutations are masked in most individuals, because the dominant gene is expressed in the phenotype. However, if two carriers for a disease both pass on the mutated allele then it is expressed in the offspring. They are called **lethal recessives**, because it will eventually cause the death of the individual they are found in.

**Extra information**

- Other mutations are called **conditional lethals**, because they only cause death if the individual with the alleles is exposed to a particular environmental factor. For example, **favism** is a sex-linked inherited condition that causes the individual to develop haemolytic anaemia (the abnormal breakdown of erythrocytes) if they eat fava beans.

| Genetic disorder    | Cause and effect                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Further information                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
|---------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Sickle-cell anaemia | <ul style="list-style-type: none"> <li>• The DNA molecule that codes for the beta amino acid chain in haemoglobin has a mutation whereby adenine is replaced by thymine. This results in replacement of one glutamic acid molecule with a valine molecule, so the haemoglobin molecule with the defect forms abnormally long fibres when the oxygen levels in the blood are low.</li> <li>• Consequently, these abnormal haemoglobin molecules cause the erythrocytes to form a crescent shape.</li> <li>• Early symptoms may include; painful swelling of the hands and feet, fatigue from anaemia and a yellowish colour of the skin, known as <i>jaundice</i>.</li> <li>• The disease is usually fatal, as sickle-cells are unable to carry as much oxygen as normal erythrocytes. Also, the cells stick together, forming clots in small blood vessels.</li> <li>• Individuals who have the sickle-cell trait but aren't double recessive suffer no ill effects where oxygen levels are normal.</li> </ul> | <ul style="list-style-type: none"> <li>• In countries where malaria is prevalent, the <b>sickle-cell trait</b> provides a selective advantage.</li> <li>• Individuals who have the allele, but also have a normal allele express <b>codominance</b> in their phenotype, with 30-40% of the erythrocytes being sickle-celled. These individuals suffer less severe anaemia and rarely die from the condition. The malarial parasite, <i>Plasmodium</i>, cannot easily invade sickle-cells.</li> <li>• Individuals with the sickle-cell trait in areas where malaria is prevalent are more likely to survive and reproduce. Thus the sickle-cell frequency is higher in these countries.</li> <li>• In countries like Australia, where malaria isn't prevalent, there is no advantage to having the sickle-cell trait, so it isn't selected for, so the frequency in the gene pool is much lower.</li> </ul> |

**Extra information****Thalassaemia ( $\alpha$  and  $\beta$ )**

- An alteration of one of the genes that produces either the alpha ( $\alpha$ ) or beta ( $\beta$ ) haemoglobin chains. Depending on how many genes are altered will determine the degree of anaemia that results.
- It is potentially fatal to young babies unless they receive a blood transfusion quickly. If they have four genes altered (two on each chromosome), they have a condition called alpha thalassaemia major or hydrops fetalis. Babies with this condition usually die before or shortly after birth.
- In Mediterranean regions it was not uncommon for cousins to marry each other, so in this gene pool, the frequency of the thalassaemia causing alleles is higher.
- In Australia, the alleles are more prevalent in those who are from Mediterranean areas, particularly immigrants from Italy and Greece and their descendants.
- People with thalassaemia require frequent blood transfusions throughout their life. Also, they may need chelation therapy, whereby they are given a chemical that binds with iron and other heavy metals to help remove the excess iron from the body.
- A bone marrow transplant may be necessary or surgery to remove the spleen or gall bladder.

### Tay-Sachs disease (TSD)

- There is a defect in one of the genes coding for the enzyme hexosaminidase A (hex A).
- This leads to a build-up of fatty proteins in the nerve cells in the brain causing blindness, deafness and reducing the ability to swallow.
- There is no cure for TSD, but pain can be relieved with medication and seizures can be managed.
- However, a baby with the condition will develop normally for the first few months, and then deterioration causing mental and physical disabilities begins. Death usually occurs in early childhood.
- Individuals of Jewish descent from Eastern Europe (the Ashkenazi Jews) have a higher frequency of the allele in their gene pool.
- Descendants from the Ashkenazi Jews can trace their roots to a group of about 330 people who lived 600-800 years ago. For many years this group only married within their faith, so there was a lack of gene flow and the presence of double recessives was higher.

## CHROMOSOMAL MUTATIONS

- Chromosomal mutations occur when more than one gene is affected and it may result in a mutation in all or part of a chromosome.
- The types of chromosomal mutations are as follows:
  - **Deletion:** a portion of a chromosome is lost. As this results in the loss of a number of genes, it usually has a significant effect on the individual's development, often proving lethal.
  - **Inversion:** a portion of chromosome becomes unattached and reattaches itself in an inverted position. The sequences of base pairs in this section of the chromosome are therefore reversed. The overall genotype is unchanged, but the phenotype is likely to be altered.
  - **Translocation:** a portion of chromosome breaks off and rejoins at a different point on the same chromosome or with a different chromosome. If it joins with a different chromosome, it is similar to crossing over, except it occurs between non-homologous chromosomes.
  - **Duplication:** a portion of chromosome is doubled, resulting in the repetition of a gene sequence.
  - **Non-disjunction:** This occurs in anaphase I or II of meiosis, when the homologous chromosomes within a pair or sister chromatids don't separate. So daughter cells either have an extra chromosome or one less. Sometimes this is referred to as **aneuploidy**, a change in chromosome number.
- As well as Down's syndrome, other examples of trisomy include:
  - **Patau syndrome:** An extra chromosome 13 results in individuals with mental retardation and a small head and sloping forehead, yet a normal birth weight. Their noses are usually bulbous and eyes are low-set and unusual in shape. A cleft lip and palate, as well as heart defects are common.
  - **Klinefelter's syndrome:** Affects about 1 in 650 men. This may result in individuals who have a genetic constitution of XXY or XYY. Sufferers suffer from infertility, low testosterone and small testes. There may be abnormal breast development and body proportions are generally female.

- Examples of monosomy include:
  - **Cri-du-chat syndrome (5p-minus syndrome):** Part of chromosome 5 is missing. Infants with this condition often have a high-pitched cry that sounds like a cat. The disorder is characterized by intellectual disability and delayed development, small head size, low birth weight, and weak muscle tone (hypotonia) in infancy. Affected individuals also have distinctive facial features, including widely set eyes (hypertelorism), low-set ears, a small jaw, and a rounded face. Some infants are born with a heart defect.
  - **Turner's syndrome:** These females have a missing X chromosome, so their **genotype** (the actual genetic information present) is XO. Often, individuals with this condition don't survive pregnancy. Those who do survive have are small in stature and sexually immature. Despite having a single X chromosome, like males, they are females. This demonstrates the importance of the Y chromosome for maleness. Appropriate medical treatment and support can help women with the condition leading a normal, healthy, productive life. Treatment is aimed at correcting any physical defects and helping to bring about puberty.



# QUESTIONS

20 marks

1. Describe what a gene pool is. [1]

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2. Outline the differences between a germline and somatic mutation. [4]

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3. What is a chromosomal mutation and describe FOUR types of chromosomal mutation. [5]

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4. Down's syndrome is a chromosomal mutation. What is Down's syndrome and explain how it occurs. [3]

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# 6

## BIOTECHNOLOGY (UNIT 4)



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                       | Done | Revise | Teach others |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Techniques in biotechnology (Unit 4)</b>                                                                                                                                                                                                                               |      |        |              |
| • I can conduct investigations, safely, competently and methodically for valid and reliable collection of data. (SIS)                                                                                                                                                     |      |        |              |
| • I understand that gene therapy can be used to treat a range of diseases, including diabetes mellitus (Unit 3). (SHE)                                                                                                                                                    |      |        |              |
| • I know that hormones and vaccines can be developed using recombinant DNA technology (Unit 3). (SHE)                                                                                                                                                                     |      |        |              |
| • I appreciate that developments in biotechnology, for example polymerase chain reaction (PCR), gel electrophoresis and DNA sequencing, have increased access to genetic information of species and provide evidence of evolution. (SHE)                                  |      |        |              |
| • I understand that developments in the fields of comparative biochemistry (including DNA and protein sequences) and bioinformatics have enabled identification of further evidence for evolutionary relationships, which help refine existing models and theories. (SHE) |      |        |              |
| • I appreciate that cell replacement therapy has the potential to treat nervous system disorders including Alzheimer's and Parkinson's diseases. (Unit 3) (SHE)                                                                                                           |      |        |              |

**Helpful hints:**

- Learn the different terms used in biotechnology, because they can get a little confusing.
- Be specific with your language, e.g. don't just write polymerase, write DNA polymerase. (RNA polymerase is for protein synthesis – *not in syllabus*).
- Often biotechnological techniques are used to answer the final part of an extended question, e.g. how are synthetic hormones made using biotechnology?

**Key points:**

- Biotechnology is the application of scientific and engineering principles to the production of materials by biological agents, which are useful to humans.
- What this chapter is really about is **recombinant DNA technology** or **genetic engineering**, a subset of biotechnology, whereby genetic material is manipulated by inserting genes from one organism into another organism in order to produce the desired products, e.g. human insulin and human growth hormone.
- The alteration of genetic material in a way that does not occur naturally by mating is done using **genetically modified organisms (GMOs)**.
- The techniques used in recombinant technology include DNA sequencing, DNA profiling, polymerase chain reaction (PCR) and gel electrophoresis.
- The applications of these techniques is to help us understand how genes function and regulate processes in the body; to give us a better understanding of evolution; for medical purposes, such as gene therapy, diagnosis of genetic disorders, and the development of vaccines and hormones; for amplifying DNA quickly for identification purposes.

**INTRODUCTION**

- Simply put biotechnology is the manipulation of living things to produce useful products.
- Several centuries ago, people manipulated genes by using selective breeding techniques to produce offspring with the desired characteristics.
- Since those early days, biotechnology has grown to include fuel production, antibiotics, hormones, waste disposal, genetically modified foods, medicines and use in forensic science.
- Much of this growth is due to a better understanding of DNA, since its structure was first published in 1953.
- In 1990, a consortium of 20 international research centres began work on sequencing the human genome. Now, we have sequenced the whole human **genome**, i.e. the complete set of genetic information of a person. The human genome project (HGP) was completed in 2003.
- Whilst the HGP simply states the order of nucleotide bases in humans, research is focussed on identifying genes in the genome, with the hope that genetic diseases can be identified, treated and prevented in the future.
- Many genes have already been identified and over 4000 genetic disorders are known. The hope is that knowing the location of these faulty genes will enable scientists to carry out gene therapy to replace them with functional genes.
- To date, the HGP has been used to develop genetic tests which check for a particular sequence of bases on the DNA to see whether they are faulty or not. Tests for haemophilia A and B, Huntington's disease, cystic fibrosis, Tay-Sachs disease and PKU, amongst others have been developed.
- Information about a person's DNA can be useful in the field of forensic science (See later notes).
- Knowing the human DNA profile raises some ethical questions, but most people think that the benefits outweigh the concerns. Now there is the possibility of improved genetic testing, New gene therapy treatments are becoming available and in the future medicine may be personalised to an individual's specific genetic needs.

- The HGP can also be very useful for the understanding of human evolution and human migration. It may help lead scientists to gain a better understanding about how humans evolved and how humans are evolving today.

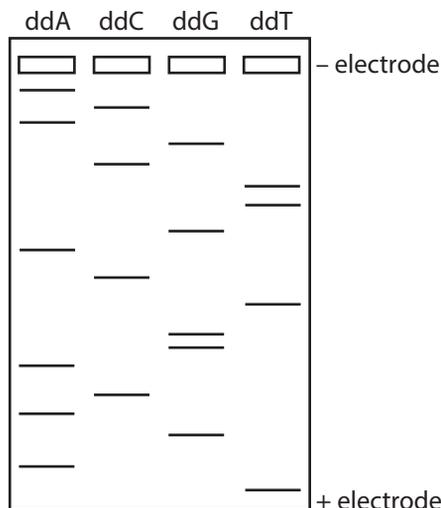
**Background information (Prior knowledge from Unit 1 and 2)**

- DNA and RNA are polynucleotides, i.e. they are large molecules made from chains of nucleotides.
- A nucleotide is composed of three parts; a pentose sugar, a phosphate group and a nitrogenous organic base.
- The nucleotide strands are made by condensation reactions (removal of water) between the phosphate and the hydroxyl on the pentose sugar. The bond is called a **phosphodiester bond**.
- The differences between the nucleic acids DNA and RNA are as follows:
  - o DNA is double stranded with each strand an equal distance apart running in opposite directions (**anti-parallel**); RNA is single stranded.
  - o DNA has the pentose sugar deoxyribose; RNA has the pentose sugar ribose. Deoxyribose has one less oxygen in it.
  - o The nitrogenous (organic) bases in DNA are: adenine (A), thymine (T), guanine (G) and cytosine (C). In RNA, thymine is replaced by uracil (U).
- In DNA, the purine (double ringed structure) adenine always pairs with the pyrimidine (single ringed structure) thymine, held together by two hydrogen bonds. Guanine (purine) always pairs with cytosine (pyrimidine), held together by three hydrogen bonds. These hydrogen bonds hold the two polynucleotide strands together to form a double helix.
- During semi-conservative replication of DNA, **DNA polymerase** is involved in pairing up complementary bases to the two strands that have been separated. It adds each nucleotide one at a time. So the two new DNA molecules contain one old strand and one new strand. This occurs during interphase, before the cell divides.

## DNA SEQUENCING

- This is the process by which the precise order of the nucleotides within a DNA molecule is determined.
- The technique was originally developed by Frederick Sanger, who won his second Nobel prize for Chemistry for his work in the 1980. His technique, called the **Sanger method** or **dideoxy sequencing**, built on the work of Ray Wu, a Chinese American biologist, based at Cornell University.
- The method used is as follows:
  - o The DNA is denatured by heating to a high temperature (96 °C) to form single stranded DNA.
  - o A primer is attached to the DNA **template** (non-coding) strand and the strand is equally distributed into four tubes. Also, DNA polymerase is added to ensure complementary DNA strands will be formed, as well as the four DNA nucleotides (dNTPs)
  - o Into each tube a different DNA nucleotide is placed, but with hydrogen synthetically replacing the hydroxyl group on the deoxyribose sugar, so, effectively, it has one less oxygen than normal, i.e. ddA, ddC, ddG, ddT. When one of these **dideoxy nucleotides** is inserted into the replicating chain, DNA sequencing stops at this point.
  - o As the process is repeated several times, the dideoxy nucleotides are inserted at different points in the DNA chain, forming different lengths of DNA strands.
  - o Using gel electrophoresis, the fragments of different lengths are separated for each of the four nucleotides. The shortest fragments travel furthest, the longest fragments travel least distance. In this way, the sequence of the DNA can be established.

- o The sequence of the bases can be determined by going from the longest strand (3' end) to the shortest strand (5') end. Then the complementary template strand can be determined.



- o In the example above, the sequence of the strand is:

3' ACAGCTTGACTGGACAGAT 5' (Coding strand)

Therefore, the template strand is:

5' TGTCGAACTGACCTGTCTA 3' (Template strand)

- DNA sequencing can be used to show whether a person has a faulty gene and whether they may be predisposed to a genetic disease. It is hoped that this will lead to personalised treatment for the specific disease identified.
- DNA sequencing can be used in maternity and paternity tests to help identify the mother or father of a child in cases of dispute. However, it is more common to use DNA profiling.

## DNA PROFILING

- DNA profiling is **not the same** as DNA sequencing.
- DNA profiling is used to identify individuals based on the banding pattern of fragments of DNA.
- As each individual has a unique banding pattern, it is sometimes known as **DNA fingerprinting**.
- A common misconception is to refer to the bands as a **barcode**. Whilst it may look like a barcode, it is not, so this should **not** be included in any answers on DNA profiling in tests or exams.
- The key elements involved in DNA profiling are use of restriction enzymes, polymerase chain reaction (PCR) and gel electrophoresis.
- Some of the uses of DNA profiling are:
  - o Maternal and paternal identification: To find out if the alleged mother or father is actually the biological mother or father of the child.
  - o Twins: It isn't always clear at birth whether twins are identical or fraternal. Identical twins will share the same DNA profile. Cells are taken from the umbilical cords of the two children.
  - o Siblings: An example would be adopted children may want to have DNA tests to make sure that alleged biological siblings are actually their blood brothers or sisters
  - o Immigration: Some visa applications may depend on proof of relatedness.

- o Forensic use: DNA testing can be used to compare the DNA profiles of suspects to offender samples. Some states, such as Victoria, allow the collection of blood and saliva samples from convicted criminals and suspects. DNA profiles are then kept on a database. The sample collected from the scene of a crime needs to contain something from which DNA can be collected, e.g. semen, hair, or blood sample.

a. Restriction enzymes

- In order to defend themselves against viruses, called **bacteriophages**, bacteria have developed defence mechanisms which enable them to cut viral DNA when there is a specific sequence of bases. This prevents the virus from multiplying and so restricts the virus' growth. Hence, the name given to these enzymes are **restriction enzymes** or **restriction endonucleases**.
- There are different restriction enzymes in different bacteria, which cut in different places.

| Name of Restriction Enzyme | Target Sequence                      | Bacterial origin                  |
|----------------------------|--------------------------------------|-----------------------------------|
| BamHI                      | 5' G   GATCC 3'<br>3' CCTAG   G 5'   | <i>Bacillus amyloliquefaciens</i> |
| EcoRI                      | 5' G   AATTC 3'<br>3' C TTAAG   G 5' | <i>Escherichia coli</i>           |
| EcoRV                      | 5' GAT   ATC 3'<br>3' CTA   TAG 5'   | <i>Escherichia coli</i>           |
| HindIII                    | 5' A   AGCTT 3'<br>3' T TCGA   A 5'  | <i>Haemophilus influenza</i>      |
| TaqI                       | 5' T   CGA 3'<br>3' A GC   T 5'      | <i>Thermus aquaticus</i>          |

- As DNA is **universal**, i.e. it is the same in all species, the restriction enzymes can be used to cut between the same sequence of bases in all organisms.
- Restriction enzymes always cut in the same place in accordance with the sequence of bases, as in the above table. Sometimes, this produces overlapping sections, called **sticky ends**. Using the same restriction enzyme, these sections of DNA can be inserted into the DNA of other organisms, because the complementary base pairs pair up.
- When the cut is straight, as is the case of EcoRV above, **blunt ends** are formed. This is not a problem, when cutting fragments for gel electrophoresis. However, when inserting a section of DNA into the DNA of another organism, often G and C bases are added artificially to create complementary sticky ends.
- When joining portions of DNA together, **DNA ligase** is used.
- In DNA profiling, the same restriction enzyme is used to cut fragments of DNA. Fragments are of different length. When put through the process of gel electrophoresis, the different fragments move through the gel at different speeds. This results in the formation of bands at different positions throughout the gel (See notes below). As different individuals have slightly different DNA, the fragments form bands in different positions in the gel. Those individuals, who are closely related have more in common than those who are less commonly related. This information is useful to us.
- Restriction enzymes are used in the manufacture of human growth hormone and insulin (more on this later).

b. Polymerase Chain Reaction (PCR)

- The purpose of using PCR is to **amplify the amount** of a sample of DNA, producing thousands of identical copies, and to do so **quickly**.
- In 1993, Kary Mullis was awarded the Nobel prize for Chemistry for developing the technique.

- There are three main parts to the process:
  - o **Denaturing:**
    - The two strands of DNA are separated by heating the DNA up to 96 °C.
    - At this temperature, human DNA polymerase is destroyed, so a more heat stable DNA is required, namely **Taq polymerase**, which comes from the bacterium *Thermus aquaticus*, which is found in hot springs.
  - o **Annealing:**
    - Once the DNA has been cooled to 55-65 °C, a **primer** is attached to each strand. This is a section of DNA with a known sequence, which attaches to a specific part of the single DNA chain.
  - o **Elongation (or extension):**
    - PCR only amplifies a certain region of the DNA, not the whole template. The primers act as a starting point for Taq polymerase to start adding complementary DNA nucleotides, one at a time, in a single direction, to the single stranded DNA. This takes place at 73 °C in a thermocycler and doubles the number of DNA molecules.
    - The process is repeated so four identical molecules of DNA are formed. The process is repeated over several cycles, generating many thousands of copies of DNA with the exact sequence of the original DNA molecule for that section. Due to the process resulting in compounding amplification, it is known as **chain reaction**.

#### c. Gel Electrophoresis

- Simply put, an electric current is passed through an agar gel to separate fragments of different sizes of DNA.
- DNA is **negatively charged**, so it moves towards the positive electrode (or anode). Larger fragments cannot travel as quickly through the gel as smaller fragments, so the smaller fragments travel further.
- Restriction enzymes cut the DNA when they come across specific sequences. Therefore, the fragments are of different lengths of nucleotides. In this way, bands are formed in the gel at different points.
- Comparisons can be made between DNA from different individuals, as long as the same restriction enzyme has been used, as stated above.
- The banding pattern formed during gel electrophoresis forms a DNA profile for that individual, which is sometimes called a **genetic fingerprint**.
- The DNA in samples of blood, hair, saliva and semen can be used to forensic science to help identify possible suspects who may have been present at a crime scene.

## RECOMBINANT DNA TECHNOLOGY

- As stated above, DNA is universal. Therefore, a restriction enzyme can be used to cut out a section of DNA in one organism and **the same** restriction enzyme can be used to open up DNA in another organism.
- As the same restriction enzyme has been used, it means that the “sticky ends” (if they are present) are complementary and so the cut out section can be inserted into another organism’s DNA. **DNA ligase** is used to stick these pieces of DNA together.
- An organism is described as **transgenic** when a gene is removed from one organism and transferred into another in such a way that the gene is expressed in the new host.
- In humans, the required gene is identified and then restriction enzymes are used to cut either side of the gene. Then this gene is inserted into yeast or, more usually, a bacterium.
- Bacteria have small circular pieces of DNA called **plasmids**. The plasmids are distinct from the main bacterial genome and can be exchanged between bacterial cells.

a. Obtaining the wanted gene

- If the amino acid sequence of the protein is known, the DNA for the gene can be worked out and the DNA synthesised in a laboratory.
- In some situations it is possible to isolate the messenger RNA (mRNA) that has been transcribed and make a copy of the DNA (cDNA) from the mRNA. This is done with insulin (See below)
- Sometimes a gene can be isolated from an entire genome. The DNA is first cut into fragments using restriction enzymes and inserted into plasmids (as described above)

b. Examples of Use of Recombinant DNA technologyi. **Producing Human Insulin**

- Messenger RNA is extracted from human pancreatic tissue.
- Complementary strands of DNA (cDNA) are produced using the viral enzyme **reverse transcriptase**.
- The single stranded cDNA is made into double stranded DNA by allowing the enzyme **DNA polymerase** to make a second strand of complementary nucleotides to those of the copy DNA.
- The human insulin gene is identified and has a bacterial regulator added and then, using the same restriction enzymes as were used to isolate the gene in the cDNA, the gene is inserted into a plasmid, using DNA ligase.
- The plasmid with the human gene is called a **vector**. (A vector is a carrier DNA molecule into which a DNA fragment containing the wanted gene can be inserted). It is cloned and inserted into a culture of plasmid-free bacteria. These bacteria take up the vectors. The bacteria multiply and, therefore, so do the vectors.
- Transcription and translation of the genes in the plasmid occurs, resulting in the production of human insulin.
- Then the product is purified to remove the other proteins and bacteria, resulting in pure human insulin.

ii. **Producing human growth hormone (hGH)**

- This hormone is produced in a similar way to insulin, using *E.coli* bacteria.
- hGH protein is made from 191 amino acids compared to 51 for insulin.
- Some of the messenger RNA was isolated from human pituitary gland tissue, whilst the rest was synthesised.
- The process then followed a similar pattern to that described for insulin. The restriction enzyme EcoRI was used to cut the plasmids and cDNA.

iii. **Producing Factor VIII**

- Factor VIII is a blood clotting factor, which is missing or in low supply in people with the disorder **haemophilia A**.
- Until recently, haemophiliacs would need to have regular blood transfusions to ensure they maintained their blood levels. This required blood from thousands of donors.
- Sadly, many haemophiliacs were exposed to risk of infection from viral diseases. This included exposure to human immunodeficiency disease (HIV) and hepatitis B. Consequently, several haemophiliacs died.
- With the advance of recombinant DNA techniques, Factor VIII can now be mass produced, with the added advantage that it isn't combined with other plasma proteins which could cause an immune response.
- Factor VIII is one of the largest molecules synthesised currently. It is cultured in mammalian cells and has been demonstrated to be highly effective in the control of excessive bleeding.

#### iv. Production of vaccines; example hepatitis B

- Hepatitis B is a viral disease that predominantly affects the liver.
- First introduced in 1986, the vaccine is a subunit vaccine containing hepatitis B surface antigen produced by recombinant DNA technology. It is safe and highly effective.
- The vaccine is cultured in yeast cells.
- It was first developed by inserting a gene from the hepatitis B virus into the cowpox virus.

## GENE THERAPY

- The purpose of this technique is to remove or replace faulty genes by inserting functional “healthy” genes.
- Once the position of genes has been identified, it may become possible to correct errors in the genes.
- The Human Genome Project has enabled us to identify about 4,000 potentially faulty genes. Repair or replacement is a complicated process; so much of the focus to date has been on single gene disorders.
- Cystic fibrosis and Huntington’s disease are two examples of diseases where gene therapy has the potential to correct the underlying cause of the disease by replacing the faulty gene with a healthy one.

#### a. Cystic Fibrosis

- In Australia, 1 in 25 people are carriers of the cystic fibrosis (CF) gene.
- Whilst carriers have no symptoms, it causes sufferers to produce mucus secretions which are too viscous, resulting in blockages of many of the main organs; such as the lungs and the pancreas. This causes recurrent infections, damage to these organs and eventually death.
- The gene which codes for a protein essential for the chloride transport has been identified on the long arm of chromosome 7. This gene is cystic fibrosis transmembrane conductance regulator (CFTR).
- Everyone has two copies of CFTR in every cell. If one is defective, chloride transport is still adequate, but if two are present, CF results.
- Microbiologists have succeeded in isolating and cloning the CFTR gene. 70% of its mutations consist of the same change to the normal DNA sequence, which is the deletion of three nucleotides, resulting in one amino acid missing in the protein chain; namely phenylalanine 508.
- **Germline therapy** and **somatic cell gene therapy** are two possible approaches to treat the disease.
- With germline therapy, the aim is to repair the gene in a fertilised egg so that the repaired gene would be copied into each daughter cell at mitosis. This would result in the elimination of the mutant gene, not only from the person receiving treatment, but also from their offspring.
- Ethical issues have been raised about whether we have the right to alter the genes of future generations. Consequently, treatment has focussed on somatic gene therapy, whereby only the affected tissues are targeted (even though the gene occurs in every cell of the patient’s body).
- Researchers modified the common cold virus to act as the vector to carry normal genes to the CFTR cells in the airways of the lungs.
- Early trials focussed on safety rather than treatment and, as such, the amount of gene transfer may have been too small to have any therapeutic benefit, or the benefits were too short-lived. Trials with alternative methods continue.

b. Huntington's Disease

- It is hoped that gene therapy will slow down or prevent the development of this single-gene disorder.
- The mutation occurs in a gene called IT15 found on chromosome 4, which results in the mutation of a protein called huntingtin. The progressive degeneration usually occurs after the age of 40 resulting in damage to nerves in the brain.
- Research has focussed on inserting a modified virus which delivers a corrective gene into brain cells. This boosts the brain's ability to defend itself against the effects of the defective huntingtin protein. Early trials on rats and primates have been encouraging.

c. Type I Diabetes

- A clever use of gene therapy would be for liver cells to take over from the pancreatic beta cells that autoimmunity destroys.
- Secondly, if it were possible, to use gene therapy to prevent the autoimmune response occurring against the antigens in the beta cells.
- Other types of pancreatic cells and liver cells would need to be stimulated to secrete insulin, from their own proinsulin genes. But they tend to do so at a continuous low level, not in the complex pattern that synchronises with the body's metabolism. Also, the cells need to be different in structure to the pancreatic beta cells, so that these aren't attacked by the autoimmunity.
- Gene therapy trials of Type I Diabetes haven't started yet.

d. Severe Combined Immunodeficiency Disease (SCID)

- This is an example of successful gene therapy.
- The gene coding for adenosine deaminase (ADA) is mutated and those with a double recessive mutation of the gene are unable to deaminate adenosine. The result of this is the death of lymphocytes, leaving sufferers with no immunity at all.
- The first clinical trial of gene therapy was at the National Institutes of Health in 1990 when a 4-year-old girl was treated. The design of this first trial did not attempt to correct the defective gene, only the T-cells. This girl is now clinically well and still has about 25% of her circulating T-cells carrying the corrected ADA gene more than 20 years after her treatment.
- After this initial clinical trial demonstrated that gene therapy could be carried out safely and that gene-corrected T-cells could survive for years and function normally, follow up trials were initiated attempting to cure children with ADA-SCID by targeting Hematopoietic stem cells (HSC) for gene correction. The results have been spectacular with most of the more than two dozen ADA-SCID patients attaining a significant long lasting increase of the T- and B-lymphocyte count and a remarkable improvement of immune function. Importantly, no episodes of serious adverse reactions or cases of leukaemia have occurred in the patients with ADA deficiency treated by gene therapy.

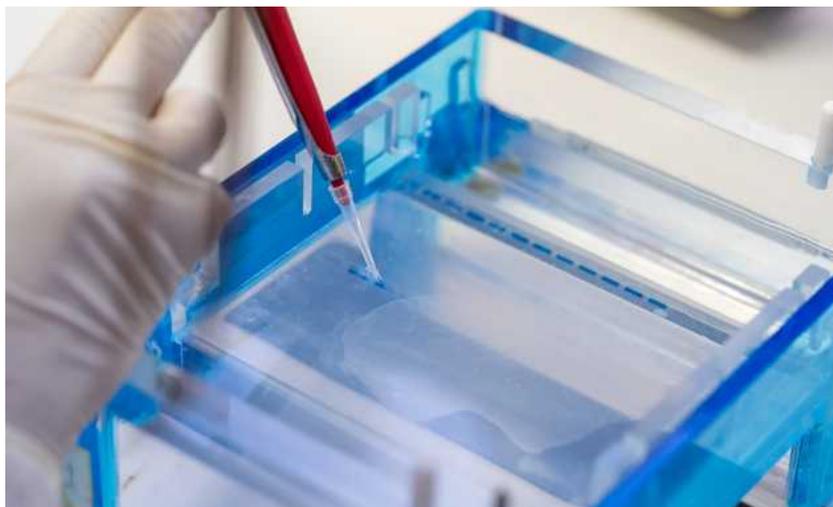
### **The possible benefits and hazards of gene therapy**

There are a number of ethical considerations to be made when gene therapy is presented as an option:

- Should somatic cell therapy, germ cell therapy or both be allowed?
- What type of patient should be considered for gene therapy?
- Are there alternative, less hazardous therapies?
- Should only life-threatening disorders be considered?
- What are the risks involved? Are there any unforeseen effects?

## CELL REPLACEMENT THERAPY AND TISSUE ENGINEERING

- **Cell replacement therapy** (also called **cellular therapy** or **cytotherapy**) is therapy in which cellular material is injected into a patient; this generally means intact, living cells.
- Two nervous system degenerative (neurodegenerative) conditions that have no known cure are Parkinson's disease (PD) and Alzheimer's disease (AD)
- AD is a form of dementia, usually occurring in people over 65 years old. Symptoms include loss of memory, followed by confusion, mood swings, aggression and general withdrawal. Use of drugs has slowed degeneration by increasing the level of neurotransmitter, acetylcholine in the brain.
- PD mainly affects older people and symptoms vary. The main ones include shaking, slow movement, muscle stiffness, stooped posture and impaired speech. Medication using drugs such as dopamine are used to relieve symptoms and manage the condition.
- If dying neuronal tissue could be replaced with healthy tissue this may cure the disease. Researchers are using induced pluripotent stem cells to grow neurones that have the same genetic background as people affected by AD or PD, allowing them to study these diseases.
- Currently, no stem cell treatments are approved for AD, but positive effects have been seen with neural stem cell transplants given to mice with a disease similar to AD. Researchers are still studying what these stem cells are doing and how they might repair nervous tissue in the brain.
- In the case of PD, current research is focussing on the pluripotent potential of various forms of stem cells. By inducing stem cells to differentiate under the correct conditions, dopaminergic neurons can be created. These neurons can be transplanted into a patient with PD, replacing their dopamine levels and providing symptomatic relief.
- **Tissue engineering** is the use of a combination of cells, engineering materials, and suitable biochemical factors to improve or replace biological functions in an effort to improve clinical procedures for the repair of damaged tissues and organs.
- An abundant supply of disease-free cells of specific types is required. Cells are induced to grow on a **scaffold**, which supports the cells until they can manufacture their own matrix structure. These scaffolds provide nutrients for the cells and are highly porous to give room for cell growth. Once established, the scaffold, together with its tissue, is implanted into the patient at the site where the new tissue is required. Eventually the scaffold degenerates leaving the new tissue.
- Currently, tissue engineering is being used with a range of tissues, including bone, cartilage, skin and adipose tissues.
- Eventually the desire is to make organs, such as kidneys, from the person's own tissue, which would not be rejected by the body's own immune system.





# QUESTIONS

20 marks

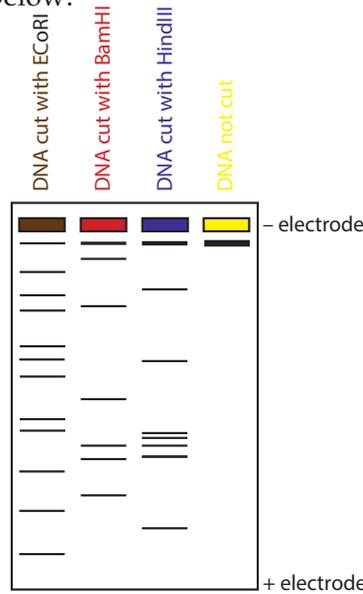
1. Explain the difference between DNA sequencing and DNA profiling? [2]

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2. Gel electrophoresis was conducted on three samples of DNA. The drawing of the sample used is shown below: [15]



- a. What is the name given to the enzymes used to cut the DNA? (1)

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- b. Why did the DNA that wasn't cut, travel the least distance? (1)

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- c. The three other sections of DNA were cut with different enzymes. Explain why the pattern of bands differs between them. (5)

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d. This DNA sample was found at a crime scene.

i. What technique would be used to amplify the amount of DNA sample?(1)

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ii. If you had a suspect for the crime, how could you use the technique named in d.i. and gel electrophoresis to identify whether the suspect is likely to have been involved in the crime? (7)

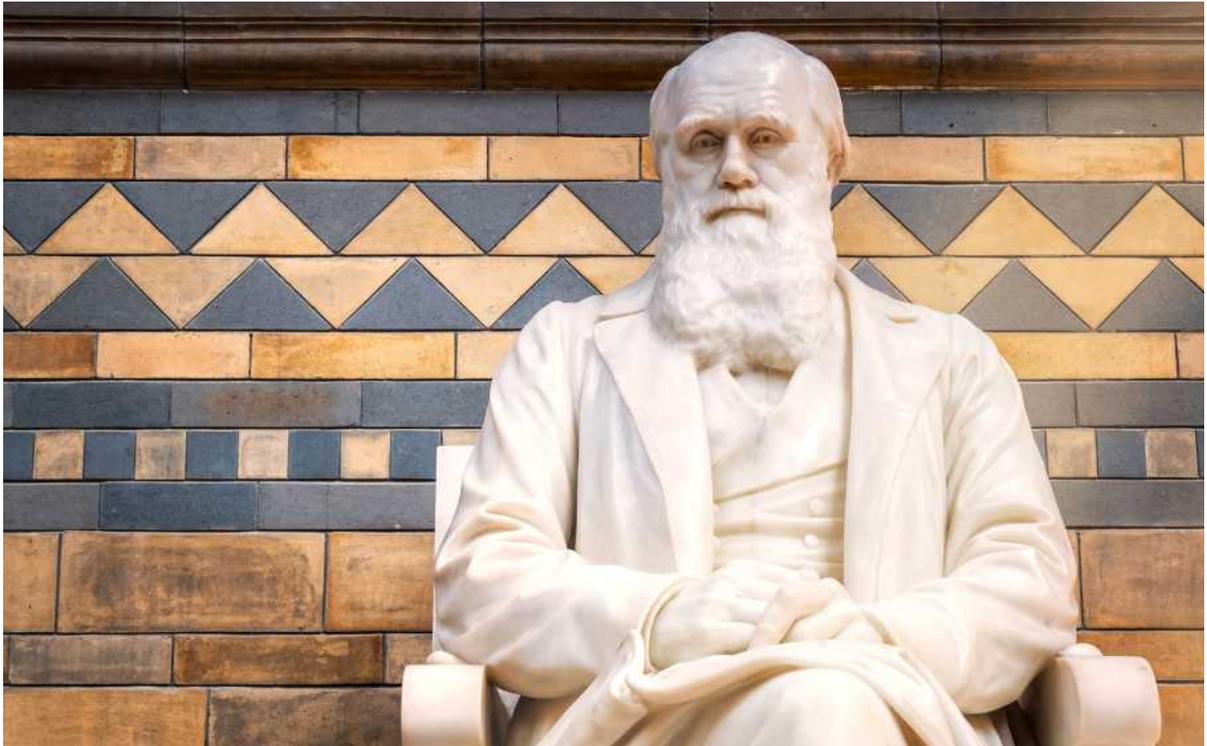
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3. What is gene therapy and how is it hoped it will be able to help people with genetic disorders, such as Huntington’s disease? [3]

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# 7

## MECHANISMS OF EVOLUTION (UNIT 4)



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                                                          | Done | Revise | Teach others |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Gene Pools (Unit 4) (Mechanisms of Evolution)</b>                                                                                                                                                                                                                                                                                                                         |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how populations can be represented as gene pools that reflect the frequency of alleles of a particular gene; gene pools can be used to compare populations at different times or locations.</li> </ul>                                                                                                                  |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how gene pools are dynamic, with changes in allele frequency caused by:               <ul style="list-style-type: none"> <li>mutations</li> <li>differing selection pressures</li> <li>random genetic drift, including the founder effect</li> <li>changes in gene flow between adjoining groups</li> </ul> </li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can discuss how the incidence of genetic diseases in particular populations illustrate the effects of different factors on the dynamics of gene pools, e.g. sickle-cell anaemia.</li> </ul>                                                                                                                                         |      |        |              |
| <ul style="list-style-type: none"> <li>I understand that natural selection occurs when factors in the environment confer a selective advantage on specific phenotypes to enhance survival and reproduction.</li> </ul>                                                                                                                                                       |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain the mechanisms underpinning the theory of evolution by natural selection include inherited variation, struggle for existence, isolation and differential selection, producing changes to gene pools to such an extent that speciation occurs.</li> </ul>                                                                |      |        |              |

**Helpful hints:**

- Recognise that mechanisms of evolution are not the same as speciation. Speciation results from mechanisms of evolution. Therefore, read the question carefully to identify if it is a question about how new species are formed, or about a specific mechanism or mechanisms of evolution.

**Key points:**

- Ensure you understand that the mechanisms of evolution differ from one another.
- Genetic drift is a random process that occurs in a small percentage of the population only.

**INTRODUCTION**

- Information is passed on from one generation to the next. This is known as **inheritance**.
- From Unit 2, we understand that sections of DNA code for particular polypeptides, which, in turn, form, structural proteins and globular proteins, or enzymes. These sections of DNA are called **genes**.
- DNA is wrapped around histone proteins, forming chromatin, which, when it condenses, forms chromosomes.
- Chromosomes pair up with their corresponding chromosome to form a **bivalent** or **homologous pair**. E.g. chromosome 17 always pairs up with chromosome 17. This means that the position or **locus** of each gene on a pair of chromosomes is in the same place.
- Sex cells, or **gametes**, are formed during meiosis, so the homologous pairs separate. Gametes only have one set of chromosomes ( $n$ ). Therefore, these cells are **haploid**.
- During sexual reproduction, a spermatozoon pairs up with an ovum to form a **zygote**. The chromosomes pair with their corresponding chromosome to form a homologous pair. As there are now two sets of chromosomes in the cell, it is **diploid** ( $2n$ ).
- For each gene, there are two pieces of information, one from the mother and one from the father. This information may be the same (**homozygous**) or different (**heterozygous**).
- If the information is different, it is usual for one gene to be expressed in the offspring (the **dominant gene** or **allele**) and one to be present, but not expressed (the **recessive gene** or **allele**).
- An allele is an alternative form of the gene.
- Sometimes, both alleles in a pair are expressed in the offspring's **phenotype**. This is called **codominance**.
- A **point mutation**, whereby a change has occurred within one nucleotide in a gene on a chromosome can be inherited. Usually this mutant gene is recessive. However, there are a few dominant point mutations. (A gene mutation is changes within a single section of DNA that codes for a polypeptide. Usually, this is a point mutation, but it could be more than one nucleotide).
- If the mutation is advantageous in some way, it is likely to be inherited by the next generation. In this way, the frequency of this allele may increase in the gene pool. Consequently, this can result in greater variation within the population.
- As most mutations are recessive, they are only expressed if both are present at a particular locus on a homologous chromosome pair, i.e. the person is double recessive for that characteristic. Often these mutations are harmful, but don't affect individuals with a normal dominant gene, i.e. the person is a carrier, but their **phenotype**, the expression of the gene, is not affected.
- When there is some effect on the genotype, it is an example of co-dominance, e.g. those individuals who have the sickle-cell trait experience some of the symptoms of sickle-cell anaemia, depending on their circumstances, but they don't experience the full disease unless they are homozygous for the trait.
- Point mutations result in variation, which can provide a selective advantage or may result in a disadvantage for individuals within a population. This is the most important mechanism by which variation can occur within a population. There are others.

## EVOLUTIONARY MECHANISMS

- **Species:** a group of individuals that share many characteristics and are capable of producing *fertile* offspring.
- Even within a species there are variations. Variations are brought about by a number of factors:
  - o Crossing over during prophase I of meiosis.
  - o Independent assortment of chromosomes/chromatids during metaphase I/II.
  - o Random choice of mate and fusion of gametes.
  - o Mutations can be point mutations or they can involve several genes on a chromosome. This second type of mutation where all or part of a chromosome is affected, is called a **chromosomal mutation**. When chromosomes fail to separate in meiosis I, it is known as **non-disjunction**, and it results in daughter cells either having one less or one additional chromosome, e.g. Trisomy 21, where an additional chromosome at position 21 results in **Downs syndrome**.
- However, even within asexual (one parent only) organisms there is variation. These variations are brought about by **mutations**. Mutations that occur within a gene are called **point** or **DNA mutations**. Such changes can be beneficial, some can be harmful, some can be benign. These **chance** occurrences can result in the development of new **alleles** (alternative forms of a gene) for a particular characteristic.
- Within a **population** (a group of organisms of the same species living together in a particular place at a particular time) there can be a variety of alleles for each characteristic. The sum of all these alleles is called a **gene pool**. Some alleles are more common than others, so we can consider the **allele frequencies** of particular alleles within a gene pool. Over time, advantageous alleles may be passed on more readily and their frequency within a population will increase. Other alleles, which are less advantageous or disadvantageous, may decline, because they are not passed on to the next generation so readily. This results in changes of the frequency of certain alleles within a gene pool.
- Mutations that occur in body cells are called **somatic mutations**. Mutations that occur in the reproductive cells are called **germline** or **germinal mutations**. Somatic mutations are not passed on, whereas germinal mutations affect the reproductive cells and can be passed on to the next and subsequent generations. (See chapter 5 for more details)

## FACTORS INFLUENCING ALLELE FREQUENCY

1. **Natural Selection:** Established by Darwin, there are 6 main factors to natural selection:
  - **Variation:** Within a population there are slight differences. Some of these differences may give some individuals within the population a selective advantage over others.
  - **Overproduction:** More individuals are produced than can be sustained by the available resources. This results in constancy of numbers, as the ecosystem can only support a certain number of individuals. If the climate changes faster than the population of a species can change, then it will die out.
  - **Struggle for existence:** Due to excessive birth rates and limited resources there is a struggle between individuals.
  - **Survival of the fittest:** Only the best adapted individuals survive to reach maturity and reproduce. Usually these individuals have alleles that give them a survival advantage, which are passed on.
  - **Like produces like:** The favourable/advantageous characteristics (those with survival value) are passed on the next generation.
  - **Over time (or over several generations) the gene pool changes:** the proportion of the favourable alleles increases and the less favourable alleles decreases.

## 2. Random genetic drift (Sewell-Wright Effect)

- In *small* populations there can be random, non-directional differences in the allele frequency which are not representative of the population as a whole. This means that in isolated populations, an allele which is rare in the population as a whole may, purely by chance, become more prevalent. E.g. 'Dunkers' from Pennsylvania didn't marry outside of their own religious community. Therefore, they constituted an isolated group within the total population of the USA. They contained allele frequencies for certain traits higher than in the gene pool as a whole.
- It is important to note that these small, isolated communities exist within the larger population, so changes in allele frequency within these gene pools are as a result of random changes between phenotypes within a population.

### Extra information

- The **bottleneck effect** is an example of what happens when a portion of the population is randomly eliminated by some, usually, catastrophic event, e.g. flooding, earthquake, volcanic eruption, destruction of habitat. In this case, those who survive may have a different frequency of alleles in the gene pool from the original population. This will result in a shift in emphasis of phenotype within the population. Therefore, the resulting population reflects the genetics of the surviving population.
- An example of the bottleneck effect occurred in 1775 on the small Micronesian island of Pingelap, where a typhoon killed nearly 90% of the population. This left only 20 survivors, one of whom was a man named Nahnmwarki Mwanenised, who had a very rare genetically inherited eye condition called achromatopsia. This recessive allele causes total colour blindness and extreme sensitivity to light. Six generations later, nearly 5% of the island's population had the condition. In the USA, only 1 in 33,000 people have the condition. Further, 30% of the population of Pingelap are carriers, whereas, in other parts of the world it is 0.0033%.

## 3. Founder effect

- This is similar to genetic drift and occurs when a group of people move away from their homeland to a totally new area and establish a new community. Again, the allele frequency for particular traits may not be representative of the population as a whole. Often in these isolated, small communities there is less genetic diversity. Such colonies may be established by major catastrophic events like a typhoon or an earthquake.
- An example of the founder effect is, the Afrikaner population of Dutch settlers in South Africa, which is descended mainly from a few colonists. Today, the Afrikaner population has an unusually high frequency of the gene that causes Huntington's disease, because many of the original Dutch colonists happened to carry that gene with unusually high frequency.
- The founder effect is easy to recognize in genetic diseases, but it must be remembered that, whilst not obvious in the phenotype, the frequencies of all sorts of genes are affected by founder events.

## 4. Migration

- Changes in allele frequency can be caused by **gene flow**, i.e. where alleles are introduced into a gene pool because of migration. One example of this is the increase of the antigen I<sup>B</sup> across Europe and Asia. Mongols have a higher proportion of this allele, but because the Mongols invaded Europe in the 12<sup>th</sup> and 13<sup>th</sup> Centuries, there was an increase in the occurrence of this allele in the rest of Asia and Europe.

## FACTORS PREVENTING GENE FLOW

- When populations are separated from each other (**isolation**) separate gene pools develop.
- There are several isolation mechanisms/barriers. These include reproductive barriers, behavioural barriers and physiological barriers. For humans, the most common barriers

are **geographical**, e.g. mountain ranges, oceans, large lake systems, deserts and expansive ice sheets. However, **sociocultural** barriers are just as effective at preventing gene flow, e.g. economic, social and educational barriers.

## GENETIC DISEASES

- Genetic diseases are the result of mutations. They cause changes to allele frequencies in a gene pool.
- Most genetic diseases are eradicated over time because people with them die before they reach reproductive age and so the allele isn't passed on. However, some alleles do persist within populations. The examples below are also included in chapter 5:
- **Sickle-Cell Anaemia:** Occurs mainly in black Africans. It is a recessive gene mutation.
  - o People who have the disease have erythrocytes that fold over into a sickle shape. Also, the erythrocytes stick to each other. The condition is fatal.
  - o Those who are heterozygous for the condition show partial effects of the disease. This is an example of **codominance**. Having the **sickle-cell trait** gives a degree of immunity to malaria. So, in these areas where malaria is prevalent, people are more prone to malaria if they don't have the sickle-cell trait. If they have the full condition, they will die. Therefore, the selection pressure in tropical areas favours those people with the partial condition who survive to reproduce and the mutant allele frequency remains high. About 40% of some populations carry the mutant allele.
  - o In areas like Australia, the sickle-cell trait is of no selective advantage and so the mutant allele is less frequent and not selected for. Therefore, the allele frequency in the gene pool is significantly less.

### *Extra information*

- **Tay-Sachs Disease (TSD):** This is a hereditary disorder of lipid metabolism.
  - o Occurs most commonly in people of Jewish descent from Eastern Europe (Ashkenazi Jews). It is caused by a missing enzyme which results in the accumulation of fatty substance in the nervous system.
  - o Death usually occurs by the age of 4 or 5. Incidence worldwide: 1 in 500,000 births. Incidence in Ashkenazi Jews: 1 in 2500 births.
  - o Two theories have been proposed: Jews often lived in isolated groups. Those who were heterozygous for the condition are less susceptible to TB. Therefore, in overcrowded, isolated conditions that would increase the threat of TB, those who got TB would die and those who got TSD would die, but carriers would survive. Therefore, the allele remains within the population. It is **selected** for by the environment, which acts as the **selective agent** (a factor that causes the death of organisms with certain characteristics, but which has no effect on individuals without those characteristics).

### *Extra information: Intelligent design and specified complexity*

- One area that is gathering momentum is the idea of intelligent design.
- Intelligent design is the theory that life and/or the universe displays such complexity that it cannot have arisen by chance, but was designed and created by some intelligent entity.
- Two ideas within intelligent design are **irreducible complexity (IC)** and **specified complexity (SC)**.
  - o IC is the idea that certain biological systems cannot evolve by successive small modifications to pre-existing functional systems through natural selection.
  - o SC states that certain patterns within biology are both specified, in that they can be described, and complex, in that it is mathematically unlikely that the event occurred by chance; it is more likely that an intelligent being is behind the specific pattern being identified.

- Intelligent design is not the same as creationism, because, according to its proponents, its aim is to look for empirical evidence to support whether the “apparent design” in nature is because of an intelligent cause or as a result of random, undirected processes. This differs from creationism, which typically starts from a point of religion and looks to seek how the findings of science can be reconciled with it.
- Intelligent design is a scientific theory, because the scientific method is applied involving the four-step process that includes observations, hypothesis, experiments and conclusion. Beginning with the observation that intelligent agents produce complex and specified information (CSI), design theorists hypothesise that for a natural object to have been designed, it would need to contain high levels of CSI. The next stage is to conduct experiments upon natural objects to see whether they do indeed contain complex and specified information. An example would be to use IC, which can be discovered experimentally by reverse-engineering biological structures. If they require all of their parts to function, e.g. the flagellum of a bacterium, then any change to this structure would render it useless. When intelligent design researchers find IC in biology, they conclude that the structures were designed. (Center for Science Culture, n.d.)<sup>1</sup>
- For more on Intelligent Design, read: “Undeniable: How Biology Confirms Our Intuition that Life is Designed” by Douglas Axe or “The Design Revolution” by William A. Dembski.

## SPECIATION

- Speciation often gets confused as a mechanism for evolution. It isn't. It is the result of evolutionary mechanisms at work.
- Speciation is the process by which new species are formed from existing species.
- The process relies on groups of individuals within one species being isolated from each other in some way. Then, based on the selection pressures at work, these groups or **demes**, tend to interbreed with individuals more often within this group than with other separate groups of the same species.
- If the flow of genes between the groups becomes less frequent, and eventually ceases, then the groups are likely to evolve along separate pathways.
- Important to recognise is that:
  - Variation within a population exists.
  - A barrier results in a separation of the population into two groups exposed to two different environmental factors. These two groups, although initially sharing the same gene pool, will eventually have separate gene pools as they are isolated from each other, i.e. no interbreeding occurs.
  - The environmental differences provide different **selection pressures** resulting in a change in gene frequency within the two separate gene pools.
  - Over time, **subspecies** develop, which are isolated but could still interbreed to produce fertile offspring if reintroduced into one group again.
  - Over a prolonged period, the changes in gene frequency may make it impossible for the two groups to produce fertile offspring. When this happens, two new species have developed. This is **speciation**.
- The two groups are likely to have the same generic (genus) name, but a different specific (species) name. This two naming system established by **Carl Linnaeus** is called the **binomial** system.
- Geographical isolation allows different groups within a species to adapt according to the environment they are in. This is known as **adaptive radiation**.

<sup>1</sup> Paraphrased from <http://www.intelligentdesign.org/whatisid.php> (Retrieved: 11.04.2017)









# 8

## EVIDENCE FOR EVOLUTION (UNIT 4)



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                | Done | Revise | Teach others |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Evidence for evolution (Unit 4)</b>                                                                                                                                                                                                                                                                                             |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain developments in biotechnology that have increased access to genetic information of species, populations and individuals, existing now or in the past, the interpretation and use of which may be open to ethical considerations. (SHE)</li> </ul>                             |      |        |              |
| <ul style="list-style-type: none"> <li>I can use information from developments in the fields of comparative biochemistry (including DNA and protein sequences) and bioinformatics have enabled identification of further evidence for evolutionary relationships, which help refine existing models and theories. (SHE)</li> </ul> |      |        |              |
| <ul style="list-style-type: none"> <li>I can describe biotechnological techniques that are used to provide evidence for evolution by using PCR (to amplify minute samples of DNA to testable amounts), bacterial enzymes and gel electrophoresis to facilitate DNA sequencing of genomes.</li> </ul>                               |      |        |              |
| <ul style="list-style-type: none"> <li>I can make comparative studies of DNA (genomic and mitochondrial), proteins and anatomy and provide additional evidence for evolution.</li> </ul>                                                                                                                                           |      |        |              |
| <ul style="list-style-type: none"> <li>I can interpret evolutionary relationships between groups that are represented by phylogenetic trees (construction of phylogenetic trees is not required).</li> </ul>                                                                                                                       |      |        |              |

| <b>Fossil evidence for evolution (Unit 4)</b>                                                                                                                                                                                                                                                  |  |  |  |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|--|--|
| <ul style="list-style-type: none"> <li>I can interpret a range of scientific and media texts, and evaluate models, processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments. (SIS)</li> </ul>                   |  |  |  |
| <ul style="list-style-type: none"> <li>I can select, use and/or construct appropriate representations, including diagrams, models and flow charts, to communicate conceptual understanding, solve problems and make predictions. (SIS)</li> </ul>                                              |  |  |  |
| <ul style="list-style-type: none"> <li>I can communicate to specific audiences, and for specific purposes, using appropriate language, nomenclature, genres and modes, including scientific reports. (SIS)</li> </ul>                                                                          |  |  |  |
| <ul style="list-style-type: none"> <li>I can explain how the fossil record is incomplete and cannot represent the entire biodiversity of a time or a location due to many factors that affect fossil formation, the persistence of fossils and accessibility to fossilised remains.</li> </ul> |  |  |  |
| <ul style="list-style-type: none"> <li>I can explain how the sequencing a fossil record requires a combination of relative and absolute dating techniques to locate fossils onto the geological time line.</li> </ul>                                                                          |  |  |  |
| <ul style="list-style-type: none"> <li>I can describe both relative dating techniques, including stratigraphy and index fossils, and absolute dating techniques, including radiocarbon dating and potassium-argon dating, and explain how each have limitations of application.</li> </ul>     |  |  |  |

### Helpful hints:

- Often contrasts are made in examination questions between different processes. Use a table to compare each contrasting point or similar point.

### Key points:

- The more features that organisms have in common, the more closely related they are and the common ancestor will be closer in the lineage. The less features the organisms have in common, the more likely that the common ancestor is further away.
- Be aware that saying that species are more or less closely related is not the same as saying they have a common ancestor. Usually, a common ancestor linking two species has not been discovered. Phylogenetic trees are used to suggest where and when in the evolutionary relationship these common ancestors could exist.
- The information collected from a variety of sources, provides a sort of portfolio to determine how closely related organisms are.
- The older an organism is, the less complex it is, the younger it is, the more complex it is.

## COMPARATIVE EVIDENCE FOR EVOLUTION

When carrying out comparative studies, the greater the similarities between different species, the more closely related the species are. The evidence gathered acts like a portfolio, whereby the different comparative studies combine to give an overall picture of how closely related, or not, different species are. The more closely related the species are, the earlier a common ancestor will be found in the evolutionary relationships.

## COMPARATIVE STUDIES IN BIOCHEMISTRY

- Comparative biochemistry is used to study the evolutionary relationships between organisms. Some examples of techniques used these evolutionary relationships is given below:

### 1. DNA

- DNA is **universal**, i.e. it is the same in all living things. However, the *sequence* of organic bases between species varies.
- New alleles are created because of mutations; others are lost by natural selection, genetic drift or other factors. This results in different proteins being made.

- Species that are more closely related have more similarities in their DNA; those which are distantly related have more differences.
- The complete set of DNA in each cell of an organism is called the **genome**. Chimpanzees share over 98% of their DNA with humans. Yet, interestingly, chimpanzees have 24 pairs of chromosomes, whilst humans have 23. It is thought that one pair of very small chromosomes has fused to another pair in humans at some time in the past.
- Studying the structure and function of similar genes in two different species is that the structure and function of a gene in one species often gives insight into its role in the other species.
- The non-coding sequences of bases in DNA (originally considered “junk DNA”, because it didn’t appear to have a function) show more similarities with more closely related species. Therefore, they have evolved from a common ancestor.
- **Endogenous retroviruses:** a viral sequence that has become part of an organism’s genome. Genetic information is stored as **RNA** not DNA. The retrovirus copies its RNA genome into DNA using a process called **reverse transcription**. A retrovirus only becomes endogenous if it becomes inserted into chromosomes that are passed on from one generation to the next. Once this happens, the ERV appears in the same place in the chromosome in every single cell and in all subsequent generations. ERVs make up to 8% of the human genome. Other primates contain some of the same ERVs in their genomes. Scientists have found 16 instances of human ERVs matching those of chimpanzees.

## 2. Mitochondrial DNA (mtDNA)

- Most DNA is found in the nucleus, but a small amount is found in the mitochondria.
- mtDNA is found in small circular molecules. Each mitochondrion has between 5-10 of these molecules.
- The genome of a mitochondrion is made up of about 16,500 base pairs.
- Within the genome, mtDNA contains 37 genes, all of which are important for normal mitochondrial functioning: 13 code for the enzymes involved in respiration, whilst 22 code for tRNA molecules and 2 code for rRNA molecules involved in protein synthesis (Some texts simply write 24 code for tRNA).
- Unlike nuclear DNA, mtDNA is inherited only from the mother, i.e. mtDNA is a copy of that found in your mother’s egg cell. This makes it easier to trace a direct genetic line.
- They have a higher rate of substitution (mutations where one nucleotide is replaced with another) than nuclear DNA making it easier to resolve differences between closely related individuals.
- Mutations occur more readily in mtDNA than in nuclear DNA. Therefore, there is a correlation between time elapsed and number of mutations that have occurred. This has allowed scientists to see how closely related individuals are by the amount of diversity in their mtDNA; the greater the diversity, the less closely related they are. One use of this technique has been to map the migration routes of ancient peoples. As a result, most Europeans are considered to have descended from hunter-gatherers who migrated to Europe in the last Ice Age, as opposed to farmers coming from the Middle East.
- Recent studies have allowed the whole Neanderthal mtDNA sequence to be completed. This has shown that Neanderthals diverged from modern humans about 600,000 years ago.

### *Extra information – Using mtDNA to establish the evolution and migration of humans<sup>1</sup>*

- Using the dataset of complete mitochondrial genomes, in a branch of science called **population genomics**, scientists have been able to provide evidence for the ‘African origin’ hypothesis. Phylogenetic trees have been constructed by determining the substitution rate of the genomic sequences. This has allowed a chronology of events to be established in the evolution and migration of early humans.
- The key date, in relation to the competing evolutionary theories, is the time when all the sequences come together to form one original sequence; the ‘mitochondrial Eve.’

- From studying the available dataset, a date of 171,500 years ago was obtained. This date is in line with that proposed in the recent African origin hypothesis.
- If multi-regionality is to be accepted, a much older date would be expected, because it would represent the common ancestor of *Homo erectus* rather than of *Homo sapiens*.

### 3. Protein Sequences

- Proteins are made from chains of amino acids; the sequence of which is determined by the genetic code on DNA.
- The degree of difference between proteins can enable scientists to determine how much time has elapsed since two species developed from a common ancestor. The longer the period of time involved, the greater the number of amino acids that would be different.
- **Ubiquitous proteins:** Found in all species from bacteria to humans. They are very basic proteins which carry out the same functions wherever they are found. By assigning each amino acid a single letter and placing them in sequence, comparisons can be made between these proteins. The number of differences in the sequence is observed; the more similarities, the more closely related they are.
  - o **Cytochrome c:** Ubiquitous protein. Performs an essential step in the production of cellular energy (part of the Electron Transfer Chain within the mitochondrion). Contains 104 amino acids, 37 of which are found in the same position in every cytochrome c molecule that has been sequenced. Scientists have found that the cytochrome c of chimpanzees and gorillas is the same as humans. Rhesus monkeys only differ by one amino acid.
  - o Other ubiquitous proteins have yielded comparable results. E.g.  $\alpha$  and  $\beta$  chains of haemoglobin are **identical** in humans and chimpanzees, but the same protein sequences in gorillas differ by one amino acid.

### 4. Bioinformatics

- Bioinformatics is the science of collecting, analysing and understanding complex biological data using computational techniques to analyse the information. It is an interdisciplinary field that combines several areas, e.g. computer science, statistics and mathematics, to interpret all types of biological data.
- It is a management information system for molecular biology.
- Some of the complex tasks include analysis of sequence information, three-dimensional structure prediction and modelling of biomolecules, examination of evolutionary relationships and biological systems, processing and analysis of biomedical images, visualization, disease research and many more.
- Once a genome has been sequenced, the sequences need to be analysed to see what they mean. This is called **genome annotation**. It involves two steps: Identifying elements of the genome (**gene prediction**), and then attaching biological information to these elements (**annotation**). Computers are used to try and analyse the information automatically.
- Initially, a tool called Basic Local Alignment Search Tool (BLAST) is used. This is an algorithm for comparing primary biological sequence information, e.g. the amino acid sequences of different proteins or the nucleotides of DNA sequences.
- Once the genes have been identified, further annotation is used to discrepancies that exist between the genomes.
- Using bioinformatics, complete genome sequences of over 40 organisms have been released ranging from organisms with genomes of 450 genes to over 100,000. Combined with data from other gene expression projects, there is a significant amount of information available for analysis. Consequently, computers have become an essential tool to handle such copious quantities of data, including the analysis and organisation of information associated with biological macromolecules.

- Comparisons of genomes and the genes they contain can be used for phylogenetic analysis, i.e. it is possible to trace the evolutionary pathways of many organisms by measuring changes in their DNA.

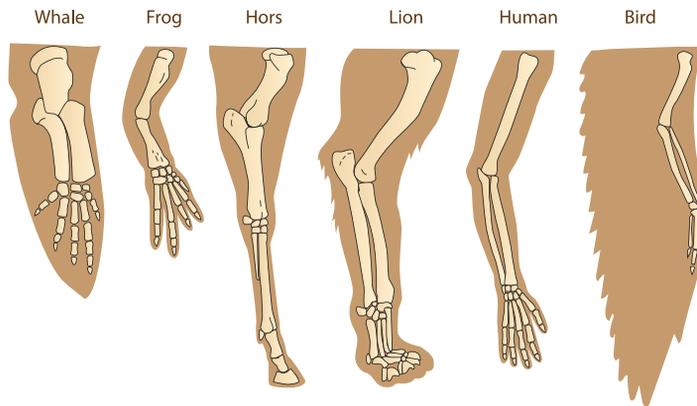
#### 5. **Comparative genomics**

- Building on the work carried out using bioinformatics, it is possible to compare the genome sequences between a wide range of organism, from bacteria to humans.
- This enables researchers to identify regions of similarities and differences, therefore providing an effective means of studying evolutionary changes among organisms.
- Genes that have been preserved can be studied, as well as those which have been altered in structure and/or their function has changed.
- Studies in comparative genomics have enabled scientists to identify subtle differences in gene sequences, which give organisms their unique characteristics, but has also shown the similarities between closely related species.
- An example of comparative genetics has revealed that humans share 60% of the genes found in a fruit fly. This suggests that the two organisms share a core set of genes.

### COMPARATIVE STUDIES IN ANATOMY

- Comparative studies are conducted to identify the similarities and differences in structures found in different organisms. These studies are often used as the first stage in studying the relatedness of species.
  - The more similarities identified, it is likely that the species have evolved from a common ancestor.
  - The more differences that exist between species suggests that the relationship between them is further apart.
  - There are three areas of comparative anatomical studies conducted:
1. **Embryology**: This is the study of the early development of an organism; in humans from fertilisation to 8<sup>th</sup> week of pregnancy.
    - Sometimes structures which are not present in the adult of the species can be seen in the embryo. This allows comparisons between organisms of different species to be made.
    - Species which are very different in adult form can be strikingly similar in their embryonic form.
    - A problem that exists in embryology is that whilst structures may have developed in a similar way, this does not necessarily mean that they came from a common ancestor. These developmental stages may have occurred independently. Therefore, often embryology is used in conjunction with evidence collected from other sources.
    - An example of comparative embryology is that the human embryo passes through a stage where it has gill structures like those of a fish. For a large portion of its development, the human embryo also possesses a tail, similar to that of its closest primates. However, usually, this tail is not present at birth. Occasionally, babies are born with this ancestral structure still intact. Tails and gills could be considered homologous structures between humans and primates, and humans and fish, but they are not present in the adults of the species.
  2. **Homologous Structures (organs)**: For some unrelated organisms, the structures have similar anatomy, but carry out different functions.
    - It is more likely that species have a common ancestor if the arrangement of bones is similarly positioned.
    - An example of this is the **pentadactyl limb**, common to all vertebrates except fish. This structure has been modified to perform distinct functions in different organisms. For example, bats and birds use it for flight, whereas in primates it forms a hand where it is used to grasp things. In whales, the limb is modified into a paddle for swimming. Though the pentadactyl limb has a variety of functions, the structures are similar, or homologous, i.e. they appear to have derived from the same common ancestor.

- Bones may be of different lengths and width, but this is the result of **adaptive radiation**, whereby the bones have been used for different purposes, so those bones best adapted for these purposes have been the ones to lead to success for that organism, resulting in evolutionary change over long periods of time.



3. **Vestigial Organs:** a structure of reduced size that appears to have no function.

- Examples include;

| Structure                    | Description                                                                                                                                         | Additional information                                                                                                                                                                        |
|------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Nictitating membrane         | A 'third eyelid'; a small fold of tissue in the corner of the eye.                                                                                  | It is called the plica semilunaris. Only one species of primate, the Calabar angwantibo, has a functioning nictitating membrane.                                                              |
| Appendix                     | At the end of the small intestine as it joins the large intestine (colon) there is an extension called the caecum, which has this blind-ended tube. | In other animals, it is larger and involved in the digestion of cellulose. Recent studies suggest that it may have a role in immune system, so it is debatable whether it is truly vestigial. |
| Coccyx                       | The final segment of the vertebral column.                                                                                                          | The remnant of a lost tail. It is debatable whether it is vestigial as it has secondary functions, such as an insertion point for some of the muscles of the pelvis.                          |
| Body hair                    | All over the body, particularly on the chest, arms and legs.                                                                                        | Unlike in most mammals, the hair on humans is too fine to create a layer of air to insulate against the cold.                                                                                 |
| Wisdom teeth                 | The third molar in the mouth.                                                                                                                       | They don't always form, but they are often removed if they do form. Ancestors used the third molar to help grind down plant material.                                                         |
| Auricular muscles of the ear | Found in the connective tissue along the side of the head behind the ear.                                                                           | The ears of humans don't move to hear potential threats. Instead, humans are capable of moving their whole head.                                                                              |

- Other structures considered vestigial include; the segmented muscles of the abdomen, nipples in males, the pyramidalis muscle (in the pelvis). Some are considered to have some function, but not all individuals have them. Therefore, they may not be considered truly vestigial.

**FOSSIL EVIDENCE FOR EVOLUTION**

- The more complex organisms of the present have all evolved from simpler organisms in the past. Scientists are looking for evidence to support these gradual changes. One major source of evidence are fossils.
- A **fossil** is any trace or remains of an organism that lived in the past.
- Its importance is that it allows scientists to get an idea of what extinct species were like. The rock surrounding fossils may provide supportive evidence as to the age of the fossil, what other organisms were around at the same time, and what the organism may have eaten.

- The hard parts of organisms that tend not to decay are bones, shells and teeth. Most fossils consist of hard parts as the soft parts decompose and the materials are recycled.

## Fossil formation

There are 6 main ways in which fossils can be formed:

1. *In shallow lakes, marshes and swamps*: the organism is quickly covered by sediment and decay stopped.
  2. *Marine habitats*: organisms are buried and preserved on the ocean floor.
  3. *Dry cave deposits*: where soft parts decay leaving hard parts undisturbed.
  4. *Trapped in ice*: low temperatures stop the decay process. Whole bodies with soft parts can be preserved.
  5. *In amber*: insects and spiders can be trapped in fossilised tree resin (not sap) and preserved intact.
  6. *Traces of organisms* include footprints, tracks, burrows, nests, larval and pupal cases and hardened dung of animals.
- The chance of fossilisation is very small due to decay by micro-organisms. However, if rapid burial occurs, conditions may not be favourable for decay. In wet, acid soils decay occurs rapidly, but in **soils with no oxygen** (and usually a low temperature) decay is slowed or prevented, e.g. in peat, so soft tissue and bone can survive.
  - **Alkaline** soils provide the best fossils, because minerals in the bone survive and deposits of new minerals, such as lime or iron oxide, are deposited in the pores of bone (**petrification**: bone is turned to rock)

## Discovery of fossils

- The main way fossils are discovered is by excavation in likely areas. A possible excavation site is surveyed and marked out into sections. Small samples are taken and sieved to ensure small fragments are not overlooked.
- **Artefacts** are objects that have been deliberately made by humans. These are often found in association with human fossils.

## Dating of fossils

- Once fossils or artefacts have been excavated in a 'dig' scientists look to determine the age of the material. There are two main **dating** techniques;
  - o **Absolute dating (sometimes called numerical dating)**: finding out the actual age of the specimen and arranging the historical specimens in order of their ages.
  - o **Relative dating**: finding out whether one sample is older or younger than another. The specimens are arranged in the geological order of their formation.
- The date of fossils or artefacts) is usually given in years before the present time, e.g. 45,000 years BP ('Before present')

### 1. **Absolute dating**

- Features of absolute dating include:
  - o It determines the age of a rock/object using radiometric techniques.
  - o Absolute dating is quantitative.
  - o This technique helps determine the exact age of the remains.
  - o It is more specific than relative dating.
  - o Absolute dating is expensive and time-consuming.
  - o It works best for igneous and metamorphic rocks.

- Absolute dating is sometimes known as **radiometric dating**, because it mainly uses the half-life of radioactive isotopes to work out the age of a fossil or a rock.
- Certain elements have **isotopes**: different forms for the same element that have a different number of neutrons in their atoms. Some of these isotopes are considered 'unstable' and can decay to different elements over time. Very unstable elements decay quickly and are considered very radioactive. Other elements take longer to decay and are considered more stable. It is impossible to measure decay of individual nuclei of the isotopes of atoms, so we measure how long it takes for half the nuclei of a particular piece of radioactive material to decay. This is called the **half-life** of the radioactive isotope.

Table to show the different types of absolute dating method (those in shaded boxes are *extra information*)

| Radiometric dating method                     | Description                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Material Used         | Useful range (BP)          |
|-----------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------|----------------------------|
| Carbon-14                                     | <p>When an organism dies, the amount of C-14 begins to decline, whereas C-12 remains stable. This process of decay takes about 5730 years to reach its half-life (<math>\pm 40</math> years).<br/>The ratio of C-14 remaining to stable C-12 can be measured.</p> <p>Problems of C-14:</p> <ul style="list-style-type: none"> <li>• Variable levels of atmospheric carbon vary with time.</li> <li>• The half-life of about 5730 years means dating is limited to 60,000 years BP.</li> <li>• Samples in the ground can interact with ground water which may contain dissolved CO<sub>2</sub> and alter C-12:C-14 ratio</li> <li>• It requires at least 3 g of organic material.<br/>This can be overcome with <b>accelerator mass spectrometry (AMS) radiocarbon dating</b>, which requires samples of 100 mg only.</li> </ul> | Carbon compounds      | Up to 60,000               |
| Potassium-argon                               | <p>The decay of potassium to argon and calcium. Potassium-40 decays to form argon-40 or calcium-40.<br/>Ratio of potassium-40 to argon-40 in sample determines age.<br/>Decay is extremely slow.<br/>Limited usefulness as not all rocks are suitable for this method of dating.<br/>Available rock needs to be of the same age as the fossil.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Volcanic deposits.    | Up to 200,000 and earlier. |
| Tree growth rings ( <b>dendrochronology</b> ) | <p>Every year trees add another growth ring of xylem (wood).<br/>Width of tree dependent on seasonal conditions that year.<br/>Core samples of wood can be used to identify conditions in the past to adjust/verify the C-14:C-12 ratio. This reduces the problem of variable atmospheric carbon levels.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | Wood                  | Up to 9000                 |
| Protoactinium                                 | <p>It has a half-life of 32,760 years.<br/>Decay product of uranium-235.<br/>Used in the dating of sediments.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Sea sediments         | Up to 250,000              |
| Uranium-thorium                               | <p>Determines the age of calcium carbonate materials such as coral.<br/>Uranium is soluble to some extent in water, whereas thorium is insoluble. As time passes, uranium-234 decays to thorium-234.</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        | Sea sediments, coral. | Up to 600,000              |

| Radiometric dating method | Description                                                                                                                                                                                                                                                                                                                                                                       | Material Used             | Useful range (BP)           |
|---------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------|-----------------------------|
| Fission tracks            | Based on analyses of the damage trails, or tracks, left by fission fragments in certain uranium-bearing minerals and glasses. Relatively simple but robust method of radiometric dating that has made a significant impact on understanding the thermal history of continental crust, the timing of volcanic events, and the source and age of different archaeological artefacts | Minerals and glass        | 100 years ago–4550 million. |
| Thermoluminescence        | The amount of luminescence is proportional to the original dose of radiation received.                                                                                                                                                                                                                                                                                            | Sediments, lava, ceramics | 300 years ago to 100,000    |

## 2. Relative dating

- Features of relative dating include:
  - It determines if an object/event is younger or older than another object/event from history.
  - Relative dating is qualitative.
  - This technique helps determine the relative age of the remains.
  - It is less specific than absolute dating.
  - Relative dating is comparatively less expensive and time-efficient.
  - It works best for sedimentary rocks having layered arrangement of sediments.
- **Stratigraphy** is the study of layers or strata. It is the oldest dating technique.
  - **Principle of superposition:** This assumes that layers at the top are younger than those beneath them. Therefore, any fossils found in the top layers are most likely to be younger than fossils found in the lower layers.
    - Distortions can occur in the Earth's crust due to moving tectonic plates, so the principle has to be applied cautiously.
    - It is possible for fossils/artefacts to be buried by animals, or early humans, sometime after deposition of sediment.
  - **Correlation of rock strata:** This is the technique of matching layers of rock from different areas.
    - Do rocks from different areas contain similar fossils if they are from the same date?
    - Wide distribution of certain fossils of limited age range is of great value in this relative dating technique. These are called **index fossils** and they are used to define geological periods, i.e. they act as guides to the age of rocks in other geographical locations. E.g. trilobite are index fossils of the Palaeozoic era.
    - The study and analysis of **fossilised pollen grain** (palynology) can help botanists to determine the type and amount of vegetation present at the time the rock layer was laid down. It gives botanists an idea of the type of vegetation that existed in those geological periods where it is found, as well as acting as index species.

### *Extra reading*

- **Fluorine dating:** When bones are left in soil, fluoride ions, which are present in the water in the soil, are absorbed by the fossils and replace some of the ions in the bone itself. All the fossil bones in a particular deposit should contain the same amount of fluoride, so that fossils that have been displaced can be detected.
- The older the fossil, the more fluoride it has.
- Fluoride concentration in ground water varies from place to place, so it is not possible to decide absolute ages.

## THE GEOLOGICAL TIME SCALE (GTS)

- This is constructed using the principle of superposition and biostratigraphic correlation (comparing similar fossils found in different locations).
- It is used by geologists, palaeontologists and other Earth scientists to describe the timing, as well as relationships, between events that have occurred in during Earth's history.
- The GTS is divided into three main categories (From largest to smallest):
  - Eras: Divided into three sections; the Cenozoic ("recent life") or Cainozoic, the Mesozoic ("middle life") and the Palaeozoic ("ancient life")
  - Periods: There are 12 periods; the most recent is the quaternary period (from 2.6 million years BP)
  - Epochs: Up to the tertiary period, these epochs are labelled generally labelled late, middle or early. These are simply sub-divisions of the periods. More specific labelling comes with the first appearance of humans.
- Humans are found in the Cainozoic era, from 65 million years BP to 0.011 (11,000) years BP, because primates first evolved in this era.

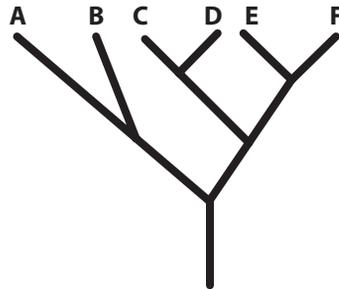
## PROBLEMS WITH THE FOSSIL RECORD

| Type of problem                                | Explanation                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
|------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Few organisms become fossils                   | Poor probability of an organism becoming a fossil. It relies on a quick burial of the material, the presence of hard body parts, an absence of decay organisms and a long period of stability.                                                                                                                                                                                                                                                              |
| Incomplete fossil record                       | Fossils need to be found. Plate tectonics, dynamic Earth movements and the cycling of rock material means many fossils have been destroyed or are located in inaccessible sites or areas yet to be explored. Also, it is unusual to find a fossil of a complete organism.<br>Human activities, such as agriculture and industry may destroy fossils or potential fossil sites.<br>Also, animals can disturb the earth through such activities as burrowing. |
| Classification of species                      | When studying extinct species it is hard to apply the principles of 'interbreeding to produce fertile offspring' to determine classification of species level.<br>Classification can depend on the number of fossils recovered to determine the range of variation for each fossil type.                                                                                                                                                                    |
| Different interpretations of the same evidence | Different scientists will use the same evidence to support different theories, e.g. the ongoing debate about the position of the Neanderthals on the human evolutionary tree.                                                                                                                                                                                                                                                                               |
| Dating methods cannot be used                  | When samples are older than 60,000 years BP then carbon-dating cannot be used. Other dating techniques rely on the material present in the sample, which, if not present, means this method of dating cannot be used.                                                                                                                                                                                                                                       |

## PHYLOGENETIC TREES (OR DENDROGRAMS)

- These diagrams reflect the historical evolutionary relationships between different organisms or species or groups of organisms (**taxa**).
- As more is discovered about different organisms, the nature of these diagrams changes.
- The end of the branches is represented by different organisms or species. The place where the branches are formed from (the nodes) are the common ancestors.
- The more closely related organisms are, the closer they feature on the phylogenetic tree. The less closely related they are, the further away from each other they are on the diagram.
- Phylogenetic trees are useful for showing the relationships between closely related organisms and showing possible or probable evolutionary pathways.

- There are limitations to them:
  - They don't necessarily accurately represent the evolutionary history of specific groups of organisms.
  - There are problems if the phylogenetic tree is based on one data set only, e.g. single gene or protein differences.
- An example of a phylogenetic tree is given below. The letters represent the current taxa.
  - C and D, and E and F has a common ancestor more recently than A and B.
  - A and B separated at the same time as there was a common ancestor for C, D, E and F.





5. What is bioinformatics and how can it be used to help understand the evolutionary relationships between organisms? [3]

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6. Researchers working on Neanderthal DNA encountered problems with both its extraction and how much survived to analyse. However, they have been successful at sequencing the mtDNA genome. [6]

- a. State the method that could be used to increase the size of the sample DNA? (1)

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- b. State the technique likely to be used to compare modern human mtDNA with that of Neanderthals? (1)

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- c. Explain how differences discovered between the modern human mtDNA and that of the Neanderthals help our understanding of evolutionary relationships? (2)

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- d. Describe two problems that the scientists may have faced in the extraction of DNA from Neanderthals. (2)

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7. Describe the difference between embryology and comparative studies of homologous structures. [2]

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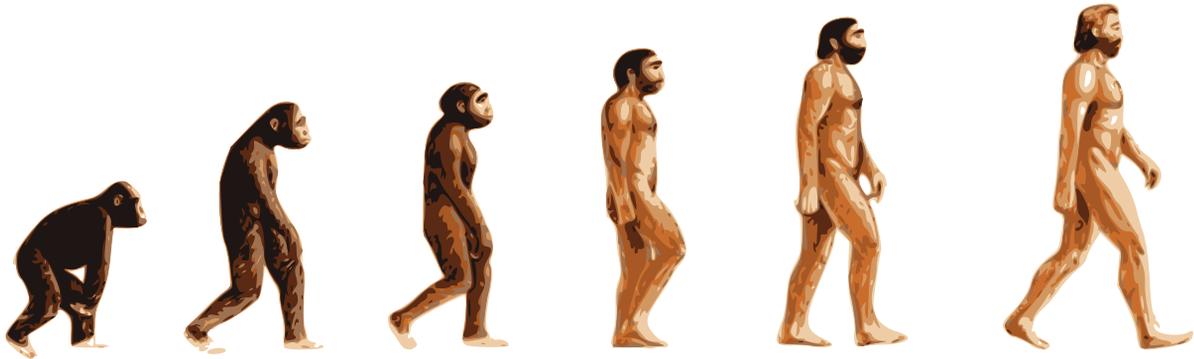
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# 9

## TRENDS AND CULTURE IN HOMINID EVOLUTION (UNIT 4)



This checklist will help ensure you cover the key points in this chapter.

| Key teaching points                                                                                                                                                                                                                                                                                                                                                                                                                                            | Done | Revise | Teach others |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------|--------|--------------|
| <b>Hominid Evolutionary Trends (Unit 4)</b>                                                                                                                                                                                                                                                                                                                                                                                                                    |      |        |              |
| <ul style="list-style-type: none"> <li>I can represent data in meaningful and useful ways; organise and analyse data to identify trends, patterns and relationships (SIS).</li> </ul>                                                                                                                                                                                                                                                                          |      |        |              |
| <ul style="list-style-type: none"> <li>I can discuss the ways in which measurement error, instrumental accuracy, the nature of the procedure and sample size may influence uncertainty and limitations in data (SIS)</li> </ul>                                                                                                                                                                                                                                |      |        |              |
| <ul style="list-style-type: none"> <li>I can select, synthesise and use evidence to make and justify conclusions. (SIS)</li> </ul>                                                                                                                                                                                                                                                                                                                             |      |        |              |
| <ul style="list-style-type: none"> <li>I can interpret a range of scientific and media texts, and evaluate models, processes, claims and conclusions by considering the quality of available evidence; and use reasoning to construct scientific arguments. (SIS)</li> </ul>                                                                                                                                                                                   |      |        |              |
| <ul style="list-style-type: none"> <li>I can select, use and/or construct appropriate representations, including phylogenetic trees, to communicate conceptual understanding, solve problems and make predictions. (SIS)</li> </ul>                                                                                                                                                                                                                            |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain that humans as primates are classified as the great apes.</li> </ul>                                                                                                                                                                                                                                                                                                                                      |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how the species within the great ape family are differentiated by DNA nucleotide sequences, which brings about differences in:                             <ul style="list-style-type: none"> <li>relative size of cerebral cortex</li> <li>mobility of the digits</li> <li>locomotion – adaptations to bipedalism and quadrupedalism</li> <li>prognathism and dentition</li> </ul> </li> </ul>           |      |        |              |
| <ul style="list-style-type: none"> <li>I can explain how determining relatedness and possible evolutionary pathways for hominids uses evidence from comparisons of modern humans and the great apes with fossils of:                             <ul style="list-style-type: none"> <li><i>Australopithecus afarensis</i></li> <li><i>Australopithecus africanus</i></li> <li><i>Paranthropus robustus</i></li> <li><i>Homo habilis</i></li> </ul> </li> </ul> |      |        |              |

|                                                                                                                                                                                             |  |  |  |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|--|--|
| ◦ <i>Homo erectus</i>                                                                                                                                                                       |  |  |  |
| ◦ <i>Homo neanderthalensis</i>                                                                                                                                                              |  |  |  |
| ◦ <i>Homo sapiens</i> .                                                                                                                                                                     |  |  |  |
| • I can discuss how tool use is seen in several hominid species and the study of these tools provides important insight into the evolution of the human cognitive abilities and lifestyles. |  |  |  |
| • I can identify and explain trends that are seen in the changes in manufacturing techniques & the materials used in the tool cultures of:                                                  |  |  |  |
| ◦ <i>Homo habilis</i>                                                                                                                                                                       |  |  |  |
| ◦ <i>Homo erectus</i>                                                                                                                                                                       |  |  |  |
| ◦ <i>Homo neanderthalensis</i>                                                                                                                                                              |  |  |  |
| ◦ <i>Homo sapiens</i>                                                                                                                                                                       |  |  |  |

### Helpful hints:

- The best way to study this module is to see it as a series of comparisons between different primates and the different hominins. E.g. The differences between apes and *Homo sapiens* or the differences between *Australopithecus afarensis* and *Homo sapiens*, or the difference between *Homo neanderthalensis* and *Homo sapiens*. This helps us to see the evolutionary trends that have occurred over time.
- When studying this topic, it may be worthwhile organising the data into tables as they are easier to read.
- It is likely that there will be an extended response question on one of the evolutionary themes, which shows the changes from quadrupedal to bipedal, or other features which can be contrasted between the great apes and modern humans.
- Ensure you can identify which group of humans were the first to use tools and fire, and identify the other key cultural milestones.
- Often there is a question involving DNA samples and fossils. DNA in fossils is in minute amounts, so PCR is used to amplify the DNA. Then gel electrophoresis is used to form DNA profiles for comparison.
- Ensure you know which type of dating can be used to date different aged fossils. E.g. Potassium-Argon cannot be used on samples over 200,000 years BP.

### Key points:

- The brain size has increased over evolutionary time with an increased cerebral cortex. The average human brain is 1350 cm
- Several changes occur within the body that enable humans to walk bipedally. This includes the reduction in size of the jaw, a change in shape of the spine from C-shape to S-shape, changes in the position of the foramen magnum to be more central, the change to the carrying angle due to the shape of the pelvis and the acetabulum, and changes to the foot creating a striding gait.
- Whilst a study of primates is not directly in the syllabus, it is necessary to understand what primates are and the differences that exist between the primates.
- Learn the key ages of hominins (Simply): *A. afarensis* (3.5 million years ago), *H. habilis* (2 million years ago), *H. erectus* (1 million years ago), *H. sapiens* (present).

## PRIMATE EVOLUTION

Humans (*Homo sapiens*), chimpanzees (*Pan troglodytes*), bonobos (*Pan paniscus*) and gorillas (*Gorilla gorilla*) are all classified at the **order** level of grouping as **primates**. Non-human primates are of interest, because they are our closest living relatives. Various sources of evidence are used to understand how humans may have evolved including; comparative anatomy of the

primates, comparative biochemistry, behaviour of living primates and fossils of primates.

Primate features (Rem: Kangaroos play some cello, orang-utans sometimes iron, panthers sing fluently, some tigers grow spots):

#### *Extra information*

- Kingdom: Animalia → animals
- Phylum: Chordata → Have vertebrates or several closely related invertebrates.
- Subphylum: Vertebrata → Have a backbone.
- Class: Mammalia → warm-blooded, produce live young, suckle their young, have fur.
- Order: Primates → Include tarsiers, lemurs, monkeys, apes and humans.
- Suborder: Haplorrhini → Include tarsiers, monkeys, apes and humans.
- Intraorder: Simiiformes → Include monkeys, apes and humans.
- Parvorder: Catarrhini → Include Old World monkeys, apes and humans.
- Superfamily: Hominoidea → Include apes and humans.
- Family: Hominidae → Include all modern and extinct orang-utans, gorillas, chimpanzees, and humans.
- Subfamily: Homininae → Include all modern and extinct chimpanzees and humans.
- Tribe: Hominini → include extinct ancestors of humans and modern humans.
- Genus: *Homo* → Some extinct ancestors of humans and modern humans.
- Species: *sapiens* → Modern humans.

- Despite all the similarities, there are many differences between primates. These differences are identified as evolutionary trends.

#### Summary of hierarchy:

Primates → Haplorrhini → Simiiformes → Hominoid → Hominid → Hominini → *Homo* → *sapiens*.

#### *Extra information on the Characteristics of Primates*

Primates are an extremely diverse group of between 190 and 350 living species. They exhibit a range of characteristic features that distinguish them from other mammals.

- **Arboreal:** living in trees. Most characteristics that primates have is because of having lived in a tree environment, e.g. grasping fingers and toes and overlapping vision. Humans have lost some features as they no longer live in trees.
- **Body:** not specialised for a particular environment.
- **Shoulders/hips:** Unlike other mammals, primates have particularly flexible shoulders and hips. Flexible shoulders allow for overarm movement, which is useful when swinging between branches. Flexible hips increase mobility, due to the greater range of motion in their legs.
- **Limbs:** generally unspecialised.
- **Hands/feet:** Pentadactyl (five fingers or toes), nails instead of claws, grasping fingers and toes with **friction ridges** (fingerprints) for gripping, first digit opposable (we have opposable thumbs). Unlike other primates, humans don't have prehensile (capable of grasping) feet.
- **Eyes:** Forward facing for stereoscopic vision. Most can distinguish colour.
- **Sense of smell:** Very poor as there is an increased focus on the use of eyes.

- **Teeth:** four incisors in both upper and lower jaw.
- **Brain:** Large and complex, cerebrum increases in size as primates become more highly evolved. The olfactory region is greatly reduced in most primates, with a greater emphasis on sight.
- **Reproduction:** Not restricted to a breeding season, rhythmical sexual cycle, usually only one offspring at a time, prolonged period of parental care for offspring.
- **Nails:** the first digit has a nail instead of claws, but most primates have nails on all fingers and toes.
- **Erect posture:** Although most primates are quadrupedal, they tend to sit or stand in a more erect posture.
- **Clavicle (collar bone):** Primates have a collarbone, not present in all mammals.

## HOMINID EVOLUTIONARY TRENDS

| Characteristic |                                                    | Trend                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
|----------------|----------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Brain          | Size                                               | <ul style="list-style-type: none"> <li>• Increasing size of brain relative to size of body.</li> <li>• Measuring the volume inside the cranium provides the <b>cranial capacity</b>, which show an increase in brain size over time.</li> <li>• Lemurs have a brain size of 24 cm<sup>3</sup>, in chimps it is 393 cm<sup>3</sup></li> <li>• Human brain: 900 cm<sup>3</sup> to 2200 cm<sup>3</sup>. Average: <b>1350 cm<sup>3</sup></b>.</li> <li>• Apes: Average = 400 cm<sup>3</sup> to 500 cm<sup>3</sup>.</li> </ul>                                                                                                                                                                                                                                                                                                                                         |
|                | Convolutions                                       | <ul style="list-style-type: none"> <li>• The shape of the brain's surface has been determined using <b>endocasts</b> (impressions of the inside of the skull left by the brain, usually made from rock or other solid material).</li> <li>• Gradual increase in the number of folds (<b>convolutions</b>) in the surface of the cerebrum. This has been worked out from endocasts.</li> <li>• Greater development of frontal lobe, which is relatively small in apes.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                  |
|                | Cerebral cortex (the outer region of the cerebrum) | <ul style="list-style-type: none"> <li>• Makes up an increasingly large portion of the brain.</li> <li>• Site of higher functions: vision, memory, reasoning and manipulative ability → development of special skills (as well as increased mobility to locate food).</li> <li>• Significant special skill: tool-making, involves a predetermined image of what completed tool should look like.</li> <li>• Changes in behaviour to meet a wide array of environmental problems.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                       |
| Digits         | Mobility                                           | <ul style="list-style-type: none"> <li>• <b>Pentadactyl:</b> 5 digits on each limb.</li> <li>• These can be moved independently of one another → increased mobility.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
|                | Opposability                                       | <ul style="list-style-type: none"> <li>• 1<sup>st</sup> digit opposable → increased effectiveness of opposability.</li> <li>• In human, no opposability in feet, because foot became weight-bearing rather than grasping appendage.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
|                | Claws/Nails                                        | <ul style="list-style-type: none"> <li>• Primitive primates retain claws on some digits.</li> <li>• Higher primates have nails on all digits.</li> <li>• <b>Friction ridges</b> (fingerprints): help increase grip between ends of digits and object. They have sense receptors in ends of fingers to help grip and manipulate objects.</li> <li>• Two types of grip: <ul style="list-style-type: none"> <li>○ <b>Power grip</b>, which enables the underside of fingers and the palm of the hand to hold onto an object tightly, whilst the thumb applies pressure in the opposite direction.</li> <li>○ <b>Precision grip</b>, which allows full manipulation of small and delicate objects using the tips of the fingers and the thumb.</li> </ul> </li> <li>• Old World monkeys are 2<sup>nd</sup> only to humans in their manipulative abilities.</li> </ul> |
|                | Prehensile                                         | <ul style="list-style-type: none"> <li>• It means capable of grasping and it is essential for climbing, allowing the ability to wrap digits around branches of trees.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |

| Characteristic                                                              |                                                                            | Trend                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
|-----------------------------------------------------------------------------|----------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Dentition                                                                   | <b>Dental formula:</b> number of each type of teeth in ¼ of the jaw.       | <ul style="list-style-type: none"> <li>Primitive mammals have dental formula: 3:1:4:3 (incisors, canines, premolars, molars).</li> <li>'Dental comb' in lemurs and lorises: used for grooming.</li> <li>36 teeth in lemurs and lorises (2:1:3:3); 32 in monkeys, apes and humans (2:1:2:3) → probably due to general reduction in jaw size (Smaller prognathism).</li> <li>Monkeys and apes: large projecting canines with diastema (gap in a row of teeth usually next to the canines in primates).</li> <li>Canines are usually longer than the other teeth.</li> <li>3-cusped molar pattern of early mammals has evolved into 4-cusp pattern in Old World monkeys. In humans and apes the lower molars have 5 cusps forming Y-5 pattern: believed to have evolved due to predominantly fruit diets of apes.</li> </ul> |
|                                                                             | <b>Dental arcade:</b> the shape made by the rows of teeth in the upper jaw | <ul style="list-style-type: none"> <li>In apes, it is much more U-shaped.</li> <li>In humans, it is more parabolic (V-shaped)</li> <li>Apes and australopithecines have larger teeth and a distinct gap between the canines and incisors. <i>Homo habilis</i> had smaller and narrower teeth, but the canines were still prominent. This tends to reflect the change in diet to softer foods, including a greater amount of meat in the diet, and eventually <b>cooked</b> food</li> <li>From <i>Homo erectus</i> to modern humans, teeth size, especially molars, have become smaller, so the jaw juts out less, leading to the more parabolic appearance.</li> </ul>                                                                                                                                                    |
| Nose                                                                        | <b>Olfaction</b> (reliance on sense of smell)                              | Reduced sense of smell with gradual reduction in size of snout. There is a reduction in prognathism (' <i>pro</i> ' (forward), ' <i>gnathos</i> ' (jaw)) in humans compared to apes, leading to a flatter face. Skulls are becoming smaller and flatter; sense of smell becomes less important when other senses, such as eyes, become more important. In apes and early Hominins, the jaw tends to protrude more.                                                                                                                                                                                                                                                                                                                                                                                                        |
|                                                                             | Protrusion of nose                                                         | Whilst the rest of the face has flattened and has less prognathism in humans, the nose has not. <ul style="list-style-type: none"> <li>In apes, who have more pronounced prognathism, they have a flattened nose.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| Extra information: To provide background on evolutionary trends in Hominins |                                                                            |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |
| Skull                                                                       | Brow ridges                                                                | <ul style="list-style-type: none"> <li>As the skull becomes larger, there are less pronounced brow ridges. So, apes have more pronounced brow ridges than humans.</li> <li>The purpose of brow ridges is to reinforce the weaker bones of the face. Therefore, they are larger in apes to cope with the extra strain put on the cranium by the jaw muscles.</li> <li>Humans have a more distinct forehead due to the reduction of the brow ridges.</li> </ul>                                                                                                                                                                                                                                                                                                                                                             |
|                                                                             | Nuchal area                                                                | <ul style="list-style-type: none"> <li>Area where muscles from the neck attach to the back of the skull, to help keep the skull balanced on the spinal cord and looking forward.</li> <li>In apes, these muscles are larger than in humans, because greater muscle strength is required with the position of the foramen magnum further forward, the C shaped spine and the larger prognathism.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                |
|                                                                             | Temporal muscles                                                           | <ul style="list-style-type: none"> <li>As these muscles help pull up the lower jaw (mandible), they are naturally larger in apes than in humans.</li> <li>Also, this relates to apes having a much more fibrous, plant-based diet, so more power is needed to grind down the food.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
|                                                                             | Zygomatic arch                                                             | <ul style="list-style-type: none"> <li>The bony arch just behind the cheeks that allows the temporal muscles to pass through.</li> <li>Naturally, these are larger in apes than in humans.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
|                                                                             | Sagittal crest                                                             | <ul style="list-style-type: none"> <li>The ridge of bone that runs along the midline of the top of the skull.</li> <li>More pronounced where there are larger jaw muscles.</li> <li>Allows attachment of one of the main chewing muscles, the temporalis fascia muscle, which is much larger in apes.</li> <li>Absent or greatly reduced in most Hominins, except the <b>Paranthropus</b> genus.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                               |

## EVOLUTION OF THE HUMAN SPECIES

- Whilst some websites refer to humans as hominids, by international convention, in terms of classification, 'family' names always end in *'-idae'* (e.g. Hominidae), 'subfamily' names end in *'-inae'* (e.g. Homininae) and 'tribe' names end in *'-ini'* (e.g. Hominini). These formal names are then abbreviated to give the common names hominid, hominin and hominini respectively.
- **Hominids** are the family group consisting of all modern and extinct Great apes, including modern humans, chimpanzees, gorillas, bonobos, orang-utans and all their immediate ancestors.
- **Hominines** are the subfamily group consisting of all modern and extinct gorillas, chimpanzees, bonobos and humans. **Pongins** are the sub-family consisting of orang-utans and their extinct ancestors.
- **Hominins** are the tribe group that consists of all modern humans, extinct human species and all our immediate ancestors, including members of the genera *Homo*, *Australopithecus*, *Paranthropus* and *Ardipethicus*).
- Whilst humans and their extinct ancestors are of the **tribe** Hominini, the other hominines are found in other tribes. Gorillas and their extinct ancestors are found in the tribe **Gorillini**; bonobos, chimpanzees and their extinct ancestors are found in the tribe **Panini**.
- Humans differ from other Homininae in appearance, structure and behaviour. They are relatively hairless and their structure allows them to be fully bipedal. Humans' erect posture and striding gait is unique. Brain size is bigger and there are changes to teeth size and shape.

### Extra information

- The closest living relatives of humans are the chimpanzee (*Pan troglodytes*) and the bonobo (*Pan paniscus*).
- Whilst they are similar in many respects to each other, bonobos and chimpanzees differ in some key social and sexual behaviours, where they show more similarity with humans than with each other.
- The study of the evolutionary relationships can be analysed by sequencing and assembling the bonobo genome, and comparing it to the human and chimpanzee genomes.
- It has been found that more than 3% of the human genome is more closely related to either the bonobo or the chimpanzee genome than they are to each other.
- This is fascinating as it allows the differing aspects of the ancestry of the two African apes to be studied and reconstructed. Also, by studying the overlapping genes, it may be possible to gain a clearer understanding of the genetic basis behind certain human phenotypes, which are shared by either the bonobo or the chimpanzee, to the exclusion of each other.

## EVOLUTIONARY TRENDS IN HOMININS

- Some of the genus *Australopithecus* evolved into the genus *Homo*.
- Human evolution: *Homo habilis* → *Homo erectus* → *Homo sapiens*. \*There were also *Homo neanderthalensis*.

## ADAPTATIONS FOR AN UPRIGHT POSTURE

A change in the skeleton and muscles of humans has occurred to allow them to walk upright, **bipedally** with a striding gait. Apes tend to maintain a **quadrupedal** (walk on all four limbs) form of locomotion:

| Structure          | Adaptation                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
|--------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Foramen magnum     | <ul style="list-style-type: none"> <li>• Located centrally in the base of the cranium, but further back in apes and earlier ancestors.</li> <li>• Evolution in a gradual move forward, so it appears on top of vertebral column. As weight of the skull is carried by vertebral column, large neck muscles are not required, whereas apes require much stronger neck muscles to keep the head forward-facing.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                               |
| Jaw bone           | <ul style="list-style-type: none"> <li>• Small and non-protruding so that it enables the skull to balance on top of the vertebral column. (Reduced prognathism over time)</li> <li>• Flatter, less protruding face means skull balances on top of spine because the weight in front of the foramen magnum is approximately equal to the weight behind.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| Chest and rib-cage | <ul style="list-style-type: none"> <li>• Broad, flatter chest (front to back), which places the centre of gravity closer to the spine.</li> <li>• Apes have a rounder, barrel-shaped chest, so the centre of gravity is further away from the spine, so they are less stable in an upright position than on all fours.</li> </ul>                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| Vertebral column   | <ul style="list-style-type: none"> <li>• Lumbar vertebrae wedge-shaped producing an S-shaped curve (double curvature) that brings the vertebral column directly under centre of skull. Contributes to upright stance. Apes have a C-shaped spine (single curvature), which forces them forward onto all fours (quadrupedal, knuckle-walkers)</li> <li>• Head balances on top of the neck due to the way the spine curves in S shape.</li> <li>• Cervical curve in neck brings vertebral column directly under the centre of gravity of the skull.</li> </ul>                                                                                                                                                                                                                                           |
| Pelvis             | <ul style="list-style-type: none"> <li>• Short and broad; shallow from top to bottom. <b>Sacrum</b> is wider, providing a wider base for support.</li> <li>• Bowl shape: Provides support for abdominal organs. In females: supports developing foetus (and usually pelvis is slightly wider).</li> <li>• Attachment of femurs wide apart contributing to carrying angle.</li> <li>• Broad hip bones provide attachments for large buttock muscles which move the legs and keep the upper body erect.</li> <li>• Broader pelvis gives stability when walking upright as weight is transferred directly to the legs.</li> <li>• A long and narrow pelvis found in apes doesn't provide the broad base of stability needed for walking bipedally.</li> </ul>                                             |
| Femurs             | <ul style="list-style-type: none"> <li>• <b>Acetabulum:</b> The socket of the pelvis in which the femur fits. Large head of femur fits into acetabulum that contributes to the carrying angle.</li> <li>• Femurs converge towards knees forming an angle to the vertical (<b>carrying angle</b>) → ensures weight distribution remains close to central axis of the body when walking. Result: greater stability, so humans have striding gait instead of swaying from side to side, as is the case in apes, who have a smaller carrying angle (Sometimes called the bicondylar angle).</li> <li>• Apes have little carrying angle, i.e. the line from hips towards the knee is almost straight and not converging inwards, resulting in less stability when standing bipedally.</li> </ul>            |
| Knee joint         | <ul style="list-style-type: none"> <li>• Outer 'hinge' larger and stronger to take weight of body, as there is greater support needed in upright stance compared to quadrupedal position.</li> <li>• Knee can be straightened.</li> <li>• Knee prevents leg from bending backwards, because ligaments in joint resist this.</li> <li>• The degree of knee movement in humans is less than in apes, because it is designed to provide strength.</li> </ul>                                                                                                                                                                                                                                                                                                                                              |
| Legs               | <ul style="list-style-type: none"> <li>• Longer than arms, especially femurs, contributing to lower centre of gravity, which contributes to stability.</li> <li>• Carrying angle allows the weight of the body to be kept close to central axis, which is not the case in apes.</li> <li>• Ankle: body weight is transmitted through the talus to the other tarsal bones, then to metatarsals and phalanges via the arches of the foot.</li> </ul>                                                                                                                                                                                                                                                                                                                                                     |
| Foot               | <ul style="list-style-type: none"> <li>• Large heel bone and aligned big toe form a pedestal on which the body is supported.</li> <li>• Foot has longitudinal and transverse arches. Transverse arch is unique to humans. Both arches allow humans to carry out perfect bipedal motion using the <b>striding gait</b> (walking in such a way that the hip and knee are fully extended). Also, the arch created can absorb energy as the foot comes down, which enables the foot to be pushed back up again, like a spring recoiling.</li> <li>• Apes only have a longitudinal arch, which makes them flat-footed and less able to maintain bipedalism for long periods.</li> <li>• Prehensibility (grasping ability) has been replaced in order to allow highly specialised bipedal motion.</li> </ul> |

## STANCE AND LOCOMOTION

- The fossil record of *Homo neanderthalensis*, *Homo erectus* and *Australopithecus* suggest they could walk upright (e.g. position of foramen magnum, shape of pelvis, structure of knee joint).
- When Hominins started to walk bipedally, with an upright body, adaptations were needed to muscles as well as bones.
- During locomotion, small changes in the contraction of muscles are needed to maintain an upright stance. These changes are controlled by **muscle tone**, the partial contraction of skeletal muscles. E.g. muscles in the back of the neck are slightly contracted to keep head upright.
- Sustained muscle tone is most evident in those muscles that support the body in an upright position, e.g. those that bring about movement of the spine, hips, knee and ankle (also abdominal muscles). The nervous system and a variety of sense organs work together to maintain the tone in these muscles.
- Striding gait: This is unique to hominins with weight transferred from heel, along outside of the foot to ball of foot. Across ball of foot to big toe. This propels the weight of the body forward. Therefore, foot is a weight-bearing appendage.
- Walking: The trunk rotates around pelvis. Arms swing to compensate for natural rotation of the body with the right arm moving forward when the left leg moves forward and vice versa. Less energy is expended as shoulders are kept at right angles to the direction of travel.
- The available evidence suggests that bipedal locomotion developed *before* any significant increase in brain size.
- The Laetoli footprints:
  - They provide evidence for hominins who walked over wet volcanic ash, which became fossilised.
  - Uncovered in 1978, these footprints believed to be over 3 million years old.
  - By this point hominins were walking upright, i.e. they were **bipedal**.
  - This form of locomotion differentiates us from apes.
  - They are believed to be made by *Australopithecus afarensis* **3.56 million years ago**.
  - Gait is not from side to side, suggesting femur is more angled than in apes, so can walk in a straighter line, as opposed to swaying from side to side (as chimpanzees today have to do).
  - The deep impression of the footprints suggest that the heel hit the ground first.
  - Although the weight distribution would not be as close to the central axis as in modern man, the footprints are more human than ape.

## THE EFFECT OF THE ENVIRONMENT ON HOMININ EVOLUTION

- Using geological and chemical information, and the types of fossilised plants and animals found, a picture can be built up of the environment at the time when ancestor hominins were alive.
- Early hominins and chimpanzees evolved from a common ancestor. Early hominins are believed to have lived in woodland or forest environments.
- Early hominins: ape-like, retained features suited to living in trees. Upright locomotion may have been used where there were gaps in the forest canopy and hominins needed to travel across open ground.
- 5-6 million years BP: Environment was changing. Temperatures dropped and forest areas declined → more open grassland areas. Natural selection would favour “apes” that were better at bipedal walking.

- Advantages of erect stance and bipedal locomotion:
  - Increased range of vision for detecting predators.
  - Increased size deterring predators.
  - Hands free for carrying food and, perhaps, for tool use.
  - Higher reach when picking fruit from trees.
  - Improved cooling of body. Upper body above ground where temperatures are slightly lower and more wind to cool body.
  - Sun would hit smaller fraction of body → less overheating.

## THE EVOLUTION OF HOMININS AND THE SEARCH FOR A COMMON ANCESTOR

- In Darwin's book *The Descent of Man*, published in 1871, he suggested that, due to their structural similarities, the great apes and man may share a common ancestor.
- Other scientists added their thoughts to this speculation, which led to a young Dutch anatomist, Eugène Dubois to go in search of fossil evidence for a common ancestor.
- His search began in 1887 unsuccessfully in Sumatra, where he went as a military surgeon and he began to excavate the caves.
- Eventually he went to Java, where he and his team discovered a jaw fragment in 1890. This was followed by the discovery of a skull cap and thigh bone.
- Dubois believed he had found the missing link between apes and humans and he published his findings in 1894. Dubois named the finding *Pithecanthropus erectus* (It became known as "Java Man").
- Later evidence identified the fossils as belonging to *Homo erectus*.
- Dubois' discovery led to the increase in for fossils.

*Table of some of the discoveries of Hominins, their average cranial capacities and year of discovery*

| Hominin                           | Cranial capacity (cm <sup>3</sup> ) | Age (millions of years BP) | Year of discovery | Early Discovery Sites                               |
|-----------------------------------|-------------------------------------|----------------------------|-------------------|-----------------------------------------------------|
| <i>Australopithecus afarensis</i> | 430                                 | Between 3.7 and 2.5        | 1974              | Hadar in Ethiopia                                   |
| <i>Australopithecus africanus</i> | 457                                 | 3.0                        | 1924              | Limestone quarry in Taung, South Africa             |
| <i>Paranthropus boisei</i> *      | 491                                 | 1.75                       | 1959              | Olduvai gorge in Tanzania                           |
| <i>Paranthropus robustus</i>      | 542                                 | Between 2 and 1            | 1938              | Kromdraai cave, Sterkfontein valley in South Africa |
| <i>Homo habilis</i>               | 590                                 | Between 2 and 1.7          | 1960              | Olduvai gorge in Tanzania                           |
| <i>Homo erectus</i>               | 1004                                | Between 1.8 and 0.03       | 1891              | Indonesian island of Java                           |
| <i>Homo neanderthalensis</i>      | 1485                                | Between 0.2 and 0.03       | 1856              | Neander valley, 18 km east of Düsseldorf, Germany   |
| <i>Homo sapiens</i>               | 1350                                | 0.2 - present              | 1868              | Cro-magnon in France                                |

\* Not in syllabus, so extra information

### *Extra information on Hominin fossil discoveries*

- After Dubois' discoveries, several paleoanthropologists looked for fossils in search of a common ancestor between apes and man.
- In 1924, Raymond Dart, an Australian anatomy professor working at the University of Witwatersrand in Johannesburg, obtained a fossil skull from the nearby limestone quarry at Taung. He named the find *Australopithecus africanus*, which literally means "southern ape from Africa."
- Robert Broom, a medical doctor, spent much of his time searching in the caves of South Africa for hominins. His most important finds were in the Sterkfontein valley. In the Kromdraai cave, he discovered fossils of a larger boned and more muscular, with a more significant sagittal crest, which is not present in *africanus* or humans. This suggests this hominin had powerful jaws. He named it *Paranthropus robustus*, meaning "parallel to man and robust."
- Between 1965 and 1983, C. K. Brain investigated the Swartkrans cave in South Africa. He discovered the remains of 130 individual hominins, believed to be the bones of australopithecines, paranthropoids and early *Homo*. Brain believed about 30 of them had used stones as tools or weapons. Others had been eaten by big cats, as one of the skulls had depressions in its head similar to the teeth of leopards.
- Louis and Mary Leakey spent time excavating in the Olduvai gorge from 1931 onwards. It wasn't until 1959 that they found their first hominin fossil, which they named *Zinjanthropus boisei*, which means "East African man" and Boisei was the name of the Leakey's benefactor. It was renamed *Paranthropus boisei*. Potassium-argon dating and stratigraphy were used to age the fossils. The Laetoli footprints were discovered by the Leakeys soon after.
- In 1974, Donald Johanson and his team of paleoanthropologists discovered "Lucy"; almost 40% of a complete adult female skeleton. This discovery was made in Hadar, in the region of Afar. It was older than Dart's discovery and was named in honour of the region, *Australopithecus afarensis*.
- Several other discoveries have been made since then, but none as complete as Lucy.

## ANATOMICAL FEATURES AND CULTURAL DEVELOPMENT OF HOMININS

### 1. Australopithecines

- Features of australopithecines are more human-like than ape-like overall, because they are considered the first Hominids to walk bipedally.
- Whilst their teeth are smaller than apes, their premolars are more like apes and they maintain a diastema.
- The carrying angle between the femur and the knee is more human-like than ape-like, indicating the bipedal form of motion.
- Many of the other features show a general trend in transition from ape-features to human-features, whereby australopithecines fall in between. E.g. they have a bigger cranial capacity than apes, but smaller than *Homo species*.
- The genus *Paranthropus* used to be known as *Australopithecus*, but it was recently changed. These are still australopithecines, but they are more **robust**, having a heavier skeleton and having maintained the sagittal crest, which suggests they have more powerful jaws than the more slender, or **gracile** species.
- *Homo sapiens* are believed to have descended from the gracile species, which has maintained the genus *Australopithecus*.
- Tools have been found with australopithecine fossils, but they are very primitive. These **pebble** or **Oldowan tools** include choppers, scrapers, flakes and chisels and have been

found at sites dating back 2.5 million years. This suggests that they were able to use the precision grip and not power grip only.

- Australopithecines lived in groups in **home bases**. Hunters and foragers went out in search of food.
- Tools allow exploitation of broader range of habitats → explore other continents and colonise them.
- There is no evidence of use of fire.

*A Table Comparing the Characteristics of Australopithecus with Paranthropus*

| Characteristic                                               | Australopithecus                    | Paranthropus                       |
|--------------------------------------------------------------|-------------------------------------|------------------------------------|
| Shallow depression in the side of the skull (temporal fossa) | Small                               | Large                              |
| Prognathism                                                  | More pronounced                     | Smaller                            |
| Sagittal crest                                               | Absent                              | Present                            |
| Forehead                                                     | Steep                               | Flat                               |
| Mandible (Jaw)                                               | Not very robust                     | Robust                             |
| Relative size of incisors and canines*                       | Large                               | Small                              |
| Relative size of premolars and molars*                       | Small                               | Large                              |
| Height                                                       | 120–140 cm (on average)/<br>Shorter | 150–170 cm (on average)/<br>Taller |
| Weight                                                       | Lighter                             | Heavier                            |

\* The teeth difference is most likely reflective of the different diets. *Paranthropus* appear to have maintained a more vegetable-based diet, which requires more grinding and chewing.

## 2. *Homo habilis* (Means “handy man”)

- Leakey and his colleagues named their discovery in the Olduvai Gorge in 1964 *Homo habilis*, because they believed that the species was the first to make tools.
- The species had the following features compared to the gracile australopithecines:
  - o It was taller, having longer femurs.
  - o It generally has a larger brain, although this is variable, and smaller teeth.
- There are problems with the labelling of *Homo habilis*, because there is so much variation between the different fossils collected, suggesting that there are several different groups that emerged from early *Homo*.
- It appears that *Homo habilis* existed alongside *Paranthropus robustus* as their fossils can often be found in the same areas.
- Also, tool-making was considered the defining feature of the genus *Homo*, but the fact that chimpanzees are adept at tool-using and other cultural behaviours, it could be that *Homo habilis* was a more advanced australopithecine.
- Despite these problems, there is certainly an advancement in development towards a more bipedal existence. *Homo habilis* does have more robust hands than modern humans, suggesting that tree-climbing was still more prominent in this species, but, with a colder and drier climate resulting in more grassland areas, more time was spent on the ground. Therefore, those better adapted to these conditions, survived and reproduced, leading to a more bipedal *Homo* species developing. Also, hands were freer than they had been when living a more arboreal existence.
- The decrease in size of teeth compared to australopithecines suggests that the diet may now have included meat. This would also account for the increase in brain size, as this high-energy food would allow the brain to develop more. Those with more developed brains would then survive to reproduce and, over time, the size of brain would become larger (According to the theory of natural selection).

- In order to survive, it is likely that hunter-gatherers would need to share food, resulting in the development of communities. Males would hunt, females would gather plant material. The diet would consist mainly of plant material, supplemented with meat.
- Due to interdependence of shared food, communication would become more important, which is likely to have led to the development of language.
- There is some evidence that early *Homo* had a bulge in the speech-producing area of the brain, but the larynx may not have been capable of making complex sounds.
- The evidence of meat eating by early *Homo* comes from animal bones found at fossil sites. Many bones show evidence of cut marks made by **stone tools** and evidence of teeth marks. *Homo* were engaged in both activities.

### 3. *Homo erectus*

- This species has a significantly larger brain than *Homo habilis* and its features are far closer to modern humans.
- Whilst *Homo erectus* was first discovered in Asia, there have been earlier forms discovered in Africa. Some paleoanthropologists label this species as *Homo ergaster*, whilst others prefer to call it "African *Homo erectus*."
- Footprints from *Homo ergaster* show the big toe parallel to the other toes, which is more similar to modern humans.
- There is evidence of fire use by this species, and increased group hunting, as well as an expansion to occupy a range of geographical areas, suggesting that the environment was no longer a selective agent on the development of hominins.
- Now *Homo erectus* was modifying the environment to suit their purposes: E.g. Use of fire, building of shelters, using a range of sophisticated tools.

#### *Extra information on Homo erectus*

- Terra Amata: Site on Riviera in France, discovered in 1966 has taught us much about the life of *H. erectus* in Europe, 40,000 years ago:
  - Huts built and there is evidence of fire and tools made from stone and bone.
  - No fossil hominins were found there.
  - There is the presence of animal bones, especially deer and a few fish.

- Other evidence suggests that *Homo erectus* were skilful hunters who employed a variety of techniques to catch prey.
- Capable of logical thought and had the ability to communicate and work with others in an organised and efficient manner.
- There seems to be a more systematic use of tools, which have been developed for specific purpose. Some tools were flaked into a hand axe. These tools are sometimes referred to as **Acheulean tools**, because the first axes were found at St. Acheul in France. Some Acheulean tools have been found with animal bones indicating their use in hunting.
- Hunting was a major source of food, but not the most important source. It is likely that hunter-gatherer practices continued.
- The importance of fire:
  - Kept predators away
  - Gave warmth and light at night.
  - Cooking became more important, which softened meat and killed parasites (and may have detoxified some plant food).
- Cultural changes: greater emphasis on cooperation, including caring for young and more complex language development.

- There are the first signs of social learning, whereby communication resulted in the development of tools. This, in turn, resulted in **collective learning**, which is the ability to retain more information with one generation than is lost by the next. Although this progress was slower in *Homo erectus*, possibly because of less communication between different groups, or just group size wasn't large enough.
- In later *Homo* species, this collective learning results in huge leaps in cultural development.

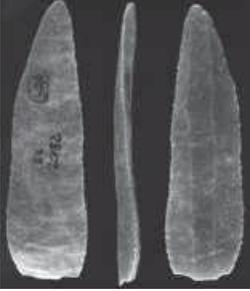
#### 4. *Homo neanderthalensis*

- About 10 km east of Düsseldorf in Germany is a valley called the Neader valley, where, in 1856, in a cave, some workmen discovered a set of bones. Whilst similar bones had been found earlier in Gibraltar, they weren't recognised at the time as being from the same species. Now, several fossils across Europe have been found belonging to what we now call *Homo neanderthalensis* (Neanderthals)
- The Neanderthals were more developed both anatomically and culturally than *Homo erectus*. They have a larger cranium, averaging 1485 cm, which is larger than *Homo sapiens*, but heavy brow ridges remain. Also, the brain was a different shape to that of humans today.
- The question that was asked was whether modern man descended from this hominid or was it a separate branch that became extinct.
- DNA samples have been extracted from Neanderthal fossils and compared to that of modern humans. This has not been an easy task as the DNA is similar and can get muddled, much has been eroded over time, and there is less of it. However, with modern techniques of PCR and gel electrophoresis it has been possible to compare the genomes, and it seems that, at some point in the past, the lineage of Neanderthals and modern man diverged.
- It is quite probable that the two species lived alongside each other in Europe for a period of time.
- Exactly why the bigger brained Neanderthal became extinct, whereas *Homo sapiens* survived, is not certain. It may be that the Cro-Magnon people, early *Homo sapiens*, either outcompeted them directly (in combat) or indirectly (through hunting and gathering the same resources) resulting in their failure to survive.
- For a period of time, they were very successful, having spread across large sections of Europe. No doubt the colder climates resulted in the development of clothes. Also, they were systematic in their use of tools and fire. Collective learning allowed Neanderthals to make advancements through tinkering, adaptation and improvement quickly with each generation and across generations.
- Tools: production of stone flakes that could be trimmed to form a variety of cutting, scraping, piercing and gouging tools. Tools were used to make other tools. There was the development of the **Mousterian industry**, named after Le Moustier in France. Tools included:
  - Flake tools enabled people living in colder climates to make clothes.
  - Scraping tools for preparing animal hides have been found at Neanderthal sites.
  - Axes with wooden handles.
- The cultural advances were not limited to tool making.
  - There is strong evidence of burying their dead, suggesting a belief in afterlife.
  - Ceremonial burials seem to have been practised.
  - Evidence of care for disabled members suggests a highly developed social system of sharing food and other resources.

## 5. *Homo sapiens*

- *Homo sapiens* have been around for about 200,000 years before present. Whilst the species has shown evolution during this time, fundamentally they are anatomically identical to present day humans. For example, they have flatter faces, larger cranial cavities, reduced or absent brow ridges, no crest on the top of the skull, smaller teeth, broader hips and longer femurs, that slope in towards the knee.
- They have adapted to be able to live comfortably almost anywhere on Earth.
- Until recently, the name **Cro-Magnon** was given to the earliest discovered *Homo sapiens* fossils in Europe, after the cave of the same name, which can be found near the village of Les Eyzies in France.
- Current scientific literature uses the term, **European early modern humans** or **EEMH**, but most people still recognise the use of Cro-Magnon.
- Uncovered in 1868, the site has been dated using Carbon-14 dating, to be about 40,000 years BP.
- Later discoveries of EEMH have been made in the same Dordogne valley, France, as well as in other parts of France, Germany and central Europe.
- The tools used by Cro-Magnon people and their counterparts around Europe were called **Aurignacian tools**, because similar tools to those found at Cro-Magnon had been discovered in Aurignac, south-western France. These tools are characterised by being formed from prepared cores rather than crude flakes. They were often made from bone as well as stone, and they date from 40,000 years to about 12,000 years ago.
- Animals provided both a source of meat, clothing and tools, as well as material for shelters.
- As well as tools, musical instruments as well as finely crafted stone and bone tools, shell and ivory jewellery, and polychrome paintings found on cave walls all testify to the cultural advancement of Cro-Magnon man.
- **Solutrean culture** was a short-lived culture that followed the Aurignacian period. It lasted from about 21,000 to 17,000 years ago in southwestern France. It was characterised by blades formed in the shape of laurel or willow leaves.
- It was succeeded by the **Magdalenian** cultural period, which was characterised by the use of bone and antlers over the use of flint and stone.
- It is believed that *Homo sapiens* demonstrated greater linguistic competence resulting in their success at colonising a range of geographical areas. Eventually this led to a move away from a nomadic lifestyle to a more village way of life. This period is often called the **Neolithic Revolution** or **Agricultural Revolution**. It is believed to have started around 12,000 years ago.
- Fig trees are believed to have been planted as long ago as 11,300 years, whilst cereals were grown in Syria 9,000 years ago. Crops, such as wheat and barley have long since been harvested. Across the world, crops were domesticated from wild varieties.
- Animal husbandry followed. Cattle, goats, sheep, and pigs all have their origins as farmed animals in the area called the **Fertile Crescent**, a region covering eastern Turkey, Iraq, and southwestern Iran. This region kick-started the Neolithic Revolution. Dates for the domestication of these animals range from between 13,000 to 10,000 years ago.
- Out of agriculture, cities and civilizations grew. Now that crops and animals could be farmed to meet demand, the global population was able to increase rapidly; from some five million people 10,000 years ago, to more than 7.5 billion today.

A table of the comparison of tools used by hominins

| Image                                                                               | Item                              | Species                          | Location found        | Tool                      |
|-------------------------------------------------------------------------------------|-----------------------------------|----------------------------------|-----------------------|---------------------------|
|    | Simple pebble tool                | <i>Homo habilis</i>              | Olduvai valley, Kenya | Oldowan pebble tool       |
|    | Hand axe                          | <i>Homo erectus</i>              | St. Acheul, France    | Acheulean tool            |
|    | Scraping tool made from flint     | <i>Homo neanderthalensis</i>     | Le Moustier, France   | Mousterian – flake tool   |
|   | Manufactured blade tool           | <i>Homo sapiens</i> (Cro-Magnon) | Aurignac, France      | Aurignacian blade tool    |
|  | Manufactured sculptured tool      | <i>Homo sapiens</i> (Cro-Magnon) | Solutré, France       | Solutrean spear heads     |
|  | Manufactured bone or antler tools | <i>Homo sapiens</i> (Cro-Magnon) | La Madeleine, France  | Magdalenian fishing tools |



# QUESTIONS

22 marks

- 1. Describe THREE changes above the pelvis that have assisted in bipedalism. [6]

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- 2. Explain the difference in tool use between australopithecines and *Homo erectus* and give an example of a tool from each species. [6]

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- 3. During an archaeological dig in volcanic rock, tools were discovered, believed to be from the Oldowan period. Name and describe the absolute-dating technique that would be used to confirm when the artefacts were from. [5]

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4. From fossil finds, archaeologists have been able to determine the cranial capacity. How have archaeologists been able to determine cranial capacity and describe the changes to cranial capacity from *Australopithecus afarensis* to *Homo sapiens* that they have observed. [5]

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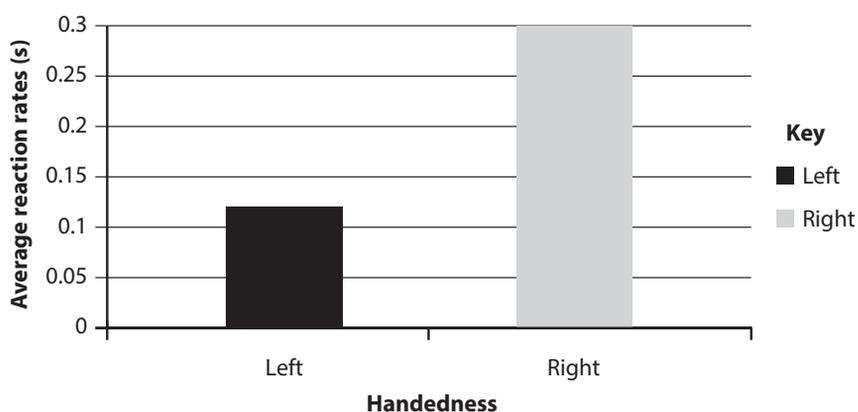


# SOLUTIONS

## Chapter 1 Scientific Inquiry (Unit 3)

- Left Handed: Total: 0.61 Average: 0.12 (There should not be a 3<sup>rd</sup> decimal place as it was recorded to 2 d.p. only) (1)  
 Right handed: 1 mark for removing 0.1 from the total. So, the total should be: 1.38 (1) (No marks for 1.48); Average: 0.3 (1) (No marks for 0.276 or 0.296 as it should be to 2 d.p. only) [3]
- The dropping of a metre ruler between the fingers on the left hand on a left-handed person will be caught more quickly than they will in the right hand on the same person OR the left hand will be better at catching the ruler after it has travelled less distance than the right hand OR a left-handed person will be better at catching a ruler in their left hand compared to their right hand (or similar) [1]
- The hand used / Left or right hand. [1]
- The distance travelled by the ruler (in cm or mm) (Units are necessary) [1]
- (Any two for one mark each):  
 He was sat down.  
 His thumb and finger were 2 cm apart before the ruler was dropped.  
 The ruler was dropped from the top of his hand each time. [2]
- He did several trials. He carried out repetition. [1]
- His hypothesis was supported (not proved). (1)  
 His left-hand was quicker at catching the ruler. (1)  
 Use of data, e.g. On average, he caught the ruler in his left hand after 0.12 seconds compared to 0.3 seconds with his right hand. (1) [3]
- His friend conducted replicates / duplicates (1)  
 This reduces the impact of biological variation and sources of error (1) [2]
- Suitable scale (1), title for graph (1), both axes labelled (1), units on the y axis (1), plotted correctly (1), independent and analysis of dependent variable on the correct axes (1) [6]

**Graph to show the average reaction rates comparing left-handed to right-handedness in a left handed person**



## Chapter 2 Homeostasis and the Nervous System (Unit 3)

1. [2]

| Sensory                                                                                                                       | Motor                                                                                              |
|-------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------|
| <ul style="list-style-type: none"> <li>Carries messages towards the CNS</li> </ul>                                            | <ul style="list-style-type: none"> <li>Carries messages away from the CNS</li> </ul>               |
| <ul style="list-style-type: none"> <li>Unipolar / Cell body on one side of axon / has one extension from cell body</li> </ul> | <ul style="list-style-type: none"> <li>Multipolar / has one axon and multiple dendrites</li> </ul> |

Each point needs to have comparative point for one mark each.

- 2.
- a. Sensory / Afferent neurone has been severed, so he can't feel his arm. (1)  
He is consciously able to send a signal to his arm via a motor / efferent neurone to move it. (1)
- b. At the end of the 1<sup>st</sup> neurone / axon terminals vesicles bind with the membrane (1)  
This releases the neurotransmitter / named neurotransmitter e.g. Acetylcholine (1) into the synaptic cleft (1)  
The neurotransmitter moves across the synaptic cleft (by diffusion) (1) and binds onto receptors on the dendrite / on the next neurone. (1)
3. [5]

|                                          | Nervous System                                                                              | Endocrine System           |
|------------------------------------------|---------------------------------------------------------------------------------------------|----------------------------|
| Nature of transmission                   | Electrochemical / Electrical <u>and</u> chemical. (Do not accept electrical on its own) (1) | Chemical / Hormonal (1)    |
| Duration of action                       | Brief / Short-lived (1)                                                                     | Longer lasting (1)         |
| Comparative speed                        | Rapid / Fast (1)                                                                            | Slower (1)                 |
| Overall effect of signal on target organ | Localised and specific (1)                                                                  | Widespread and general (1) |

4. Cranium / Bone (Do not accept skull) (1), Cerebrospinal fluid (1), Meninges (1) (In any order) [3]
- 5.
- a. B – Medulla (oblongata) (1), C – Cerebellum (1) [2]
- b. B [1]
- c. A [1]
- d. Convolutions / Gyri (1) Increase the surface area of the brain (1) [2]
- 6.
- a. Reflex [1]
- b. Unconscious / Involuntary (Do not accept "misses out the brain") (1), Rapid / Quick (1) [2]
- c. Stereotypical [1]

## Chapter 3 Homeostasis and the Endocrine System (Unit 3)

1. [8]

| Amine-based hormone                                                                    | Steroid-based hormone                                                                            |
|----------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------|
| 1. Binds to receptors on cell surface membrane / plasma membrane (1)                   | 2. Passes through the cell surface membrane / plasma membrane (1)                                |
| 3. Doesn't bind with anything other than the receptor on the cell surface membrane (1) | 4. May bind with something in the cytoplasm to form a complex (1)                                |
| 5. Causes a secondary messenger to be released. (1)                                    | 6. Either enters nucleus or enters another organelle (1)                                         |
| 7. Usually results in activation of specific enzymes (1)                               | 8. Switches gene on / Modifies gene activity / Stimulates transcription or protein synthesis (1) |

The way the question is worded means that each dot point allows the difference between the modes of action of each type of hormone to be compared. If one mode of action is described and then the other, then it isn't truly answering the question, as it is worded.

2. [6]
- Hypothalamus is joined to the pituitary gland via the infundibulum. (1)
  - Anterior lobe of the pituitary gland (ALPG) is joined to the hypothalamus via a network of blood capillaries. (1)
  - Posterior lobe of the pituitary gland (PLPG) is joined to the hypothalamus via nerve cell extensions. (1)
  - Stimulating and releasing factors control the release of hormones from the ALPG via the blood capillaries. (1)
  - Hormones stored in the PLPG are made in the hypothalamus. (1)
  - Their release is stimulated by nerve impulses in the hypothalamus which pass down the nerve extensions. (1)
3. [10]
- a. [6]

| Hormone                    | Endocrine Gland                   | Target Organ          |
|----------------------------|-----------------------------------|-----------------------|
| ----                       | Posterior lobe of pituitary gland | Kidneys (A: nephrons) |
| Adrenaline / Noradrenaline | ----                              | Heart                 |
| ----                       | Adrenal cortex                    | Kidney                |

1 mark for each answer.

- b. Acts on the kidney to reduce the amount of sodium and increase the amount of potassium in the urine. (1)
- c. Cortisol (1);
- promotes normal metabolism (1),
  - helps the body withstand stress (1) OR helps with the repair of damaged cells (1). (3)
4. Exocrine: A gland that secretes substances onto the epithelial surface via a duct. (1) [3]  
Endocrine: A ductless gland, that secretes hormones directly into the surrounding tissue / bloodstream (1)  
Example: Pancreas / Testes. (1)

5. [8]
- a. *Hyperthyroidism (Reject: Graves' Disease – this affects younger women usually)* (1)
- b. *Any three from; excessive sweating, rapid heart rate, increased appetite, weight loss, increased appetite, palpitations, insomnia, heat intolerance, difficulty falling asleep.* (4)
- c. *Surgery to remove part of/all the thyroid gland OR radioactive iodine drink to kill thyroid cells.* (1)
- d. *Suffering from hypothyroidism (1); Need extra iodine in the diet OR tablets containing thyroid hormone (1)* (2)
6. [25]
- a. (10)
- *Osmotic pressure increases / Water concentration of blood plasma decreases (1)*
  - *Detected by Osmoreceptors (1) in the thirst centre (1) of the hypothalamus (1)*
  - *Person feels thirsty (1)*
  - *(After the match) players drink large volumes of water (1)*
  - *This is absorbed into the blood from the alimentary canal / large intestine / colon (1)*
  - *Osmotic pressure decreases / Water level in the blood increases (1)*
  - *Extracellular and intercellular fluids (begin to) return to normal (1)*
  - *This is an example of negative feedback (1)*
- Any 8 points for each dot point one mark each* (6)
- b. (6)
- *Osmoreceptors in the hypothalamus detect the increased osmotic pressure / decreased water concentration in the blood plasma. (1)*
  - *Posterior lobe of the pituitary gland is stimulated (1) to release more ADH (1)*
  - *Permeability of the distal convoluted tubules and collecting ducts (1) in the nephrons of the kidney increase (1)*
  - *More water is reabsorbed into the blood plasma (1)*
  - *Osmotic pressure decreases / Water level in the blood increase (NO MARK)*
- c. (7)
- *Low blood glucose concentration is detected (1)*
  - *By pancreatic islets / Islets of Langerhans (1)*
  - *Alpha cell ( $\alpha$  cells) (1)*
  - *Release glucagon (1)*
  - *Glycogen is broken down into glucose / glycogenolysis occurs (1)*
  - *Fats and amino acids are converted to glucose / gluconeogenesis occurs (1)*
  - *Blood glucose concentration increases (1)*
- d. (2)
- *Decreasing / going down. (1)*
  - *Being used up in respiration (1)*

## Chapter 4

### Response to Infection (Unit 3)

1. [4]
- a. *Pathogen: a disease-causing organism.* (1)
- b. *Bacteriophage: a virus that infects bacteria.* (1)
- c. *Cilia: Hair-like projections from a cell.* (1)
- d. *Lysozyme: an enzyme that kills bacteria.* (1)
2. [2]
- *Non-specific: works against all pathogens.* (1)
  - *Specific: directed at a particular pathogen.* (1)
3. [3]
- *Sneezing: a sudden, involuntary expulsion caused by an irritation of the walls of the nasal cavity.* (1)
  - *Coughing: irritation in the lower respiratory tract causes a rapid expulsion of air from the lungs.* (1)
  - *Vomiting: involuntary, forceful expulsion of the contents of the stomach, caused by contractions of the abdomen and the diaphragm.* (1)
  - *Diarrhoea: irritation of the small and large intestines by micro-organisms causes contractions of the intestine walls, faster than water can be absorbed. This results in watery faeces.* (1)
- Any three descriptions for one mark each.*
4. [10]
- *The cut caused an inflammatory response (1)*
  - *due to the damage to the tissues (caused by the cut) (1)*
  - *Histamine was released (at the site of injury) (1)*
  - *which increased blood flow (1) causing redness (1) and increased heat (1)*
  - *and made the walls of the capillaries more permeable (1)*
  - *so fluid flowed into the tissue, causing swelling. (1)*
  - *Pain is caused due to stimulation of the pain receptors in the damaged area. (1)*
  - *Heparin is the other useful substance released (which prevents clotting in the immediate area of the injury) (1)*
5. [20]
- a. *Artificial, active (in either order, but BOTH needed)* (1)
- b. *Living attenuated* (1)
- c. (16)
- *Tuberculosis antigen reaches lymphoid tissue (1)*
  - *Antigen presentation (1)*
  - *B-cells / B-lymphocytes are stimulated to undergo cell division / mitosis (1)*
  - *Most B-cells develop into plasma cells (1)*
  - *which produce antibodies specific to the tuberculosis bacterium (1)*
  - *Some become memory B-cells (1)*

- They enter the blood plasma / bloodstream and lymph (1)
- On reaching the bacteria, they inactivate it by:
  - Causing bacteria to clump together / agglutination (1)
  - Coating the bacteria so they are more easily consumed by phagocytes (1)
  - Forming an antigen-antibody complex. (1)
  - Reacting with soluble antigens to make them soluble. (1)

(Any two for 1 mark each)

- Certain T-cells / T-lymphocytes undergo cell division / mitosis (1)
- Most T-cells form killer T-cells (1)
- which attach to the invading bacteria and destroy them (1)
- or form helper T-cells (1)
- which cause lymphocytes at the site of infection to become sensitised (1)
- and attract macrophages to carry out phagocytosis (1).
- Some become memory T-cells (1)
- Both T- and B-memory cells reduce the chance of a secondary response. (1)

(16 points for 16 marks, but at least 6 marks each need to come from both B-cells and T-cells)

d. Bacteria (1)

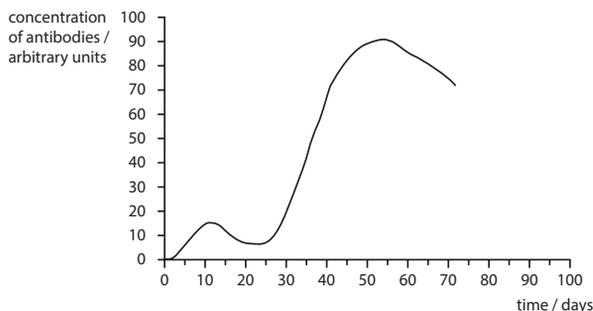
e. Antibiotics (1)

6. (6)

a.

- It took time for:
    - antigens to reach the lymphoid tissue for antigen presentation/AW (1)
    - for the plasma cells to form by mitosis or for cell division to occur. (1)
    - for the production of antibodies to occur (1)
  - There are no memory cells for a quicker secondary response. (1)
- (Any two points for one mark each)

b.



- rise starts between around day 25 (1)
  - rise is steeper and rises higher than first response (1)
  - concentration declines more slowly/with less steep gradient than primary response (1)
- (Maximum of two marks)

c. (2)

- On first exposure, there were no memory cells, so it took time to fight disease
- On secondary exposure, memory cells led to a much faster immune response, so no symptoms were experienced.

## Chapter 5

### Mutations and Gene Pools (Unit 4)

1. A sum of the alleles in a given population. [1]

2. [4]

| Germline mutation                             | Somatic mutation                                                                                  |
|-----------------------------------------------|---------------------------------------------------------------------------------------------------|
| • Affects the reproductive cells (1)          | • Affects the body cells (1)                                                                      |
| • Can be passed on to the next generation (1) | • Only the individual is affected by the mutation / Can't be passed on to the next generation (1) |

3. [5]

- Chromosomal mutation affects all or part of a chromosome, i.e. more than one gene is affected. (1)
- Any FOUR from the following for one mark each: (4)
- Deletion: part of a chromosome is deleted or lost, resulting in loss of genes.
- Duplication: a portion of chromosome is doubled, resulting in repetition in gene sequence.
- Translocation: part of a chromosome breaks off and rejoins at a different point on the same chromosome.
- Inversion: part of a chromosome breaks off and reattaches in the same position but the wrong way around.
- Non-disjunction: during anaphase I of meiosis, a homologous pair does not separate, so one of the daughter cells has an extra chromosome, whilst the other daughter cell has one less than the normal number.

4. [3]

- Trisomy 21 / An extra 21st chromosome. (1)
- Caused by non-disjunction during meiosis I. (1)
- When gametes fuse, there are 47 chromosomes in the embryo (not 46). (1)

5. [7]

- A point mutation is a change to a single gene. (1)
- Alleles are alternative forms of those genes. (1)
- When a mutation occurs in a gene, it can result in the formation of a new allele. (1)
- Over time, this can change the frequency of this gene within the population. (1)
- If the allele is favourable, it may be selected for. (1)
- This is likely to result in an increase in this allele in the population. (1)
- Therefore, there would be a change in allele frequency within the gene pool of a population. (1)

## Chapter 6 Biotechnology (Unit 4)

1. [2]
  - DNA sequencing: a process used to determine the precise order of nucleotides within a DNA molecule. (1)
  - DNA profiling: a technique used to identify the banding patterns of DNA fragments to identify particular individuals. (Sometimes called DNA fingerprinting). (1)
2. [15]
  - a. Restriction (enzymes) (1)
  - b. It is the largest fragment, so can't move/migrate through the gel as easily as smaller fragments (therefore, it doesn't travel as far in the time allocated for the electrophoresis to run). (1)
  - c. (5)
    - Restriction enzymes always cut the DNA at a point where there is a specific sequence of bases.
    - This is known as a recognition site.
    - Different restriction enzymes cut at different recognition sites / cut after specific sequences of bases,
    - forming different lengths / fragments of DNA.
    - Therefore, the sequence of bands is different (for each restriction enzyme).

(Each dot point worth one mark each, but answers need to be logically sequenced, i.e. the last mark can't be given without some understanding of the previous points being demonstrated).
  - d.
    - i. Polymerase chain reaction (Don't accept PCR; term hasn't been used earlier in the question) (1)
    - ii. (7)
      - PCR: amplifies the DNA / increases the amount of DNA (in the samples) (1), quickly (1)
      - Use a restriction enzyme on the DNA sample from the crime scene. (1)
      - Use the same restriction enzyme on a sample of DNA from the suspect. (1)
      - Use gel electrophoresis to compare the samples. (1)
      - If the bands are identical in the samples it is likely that the suspect was involved in the crime. (1)
      - Repeat the technique using a different restriction enzyme to confirm the results. (1)
3. [3]
  - Gene therapy: the introduction of a normal gene to treat / prevent a genetic disorder. (AW) (1)
  - By inserting the healthy gene, it is hoped it will replace the effects of the faulty gene. (1)

- Returning normal functioning to the person which has been affected by the faulty gene. (1)

## Chapter 7 Mechanisms of Evolution (Unit 4)

1. [5]
  - a. Founder effect. (1)
  - b. Gene / Point mutation (1)
  - c.
    - Gene flow / migration (1)
    - Resulting in introduction of new alleles. (1)
    - Therefore, the frequency of existing alleles is reduced (slightly), over time. (1)
2. [10]
  - Variation exists within a population. (1)
  - There is a shared gene pool. (1)
  - A mountain range, the ocean, a river (Accept: any geographical boundary) separates the population into two or more groups. (1)
  - This creates separate gene pools. (1)
  - Different selection pressures / selection agents act on the different groups. (1)
  - This brings about a change of allele frequency (1)
    - over time / over several generations. (1)
    - Initially, sub-species are formed (which can still interbreed). (1)
    - Over a long period of time / many generations, the groups can no longer interbreed (to produce fertile offspring) (1)
    - Two (or more) species are formed. (1)

(Each point needs to be logically structured, e.g. no marks for last point unqualified)
3. [3]
  - a. The total number of alleles in a particular population (at a particular time). (1)
  - b. The number of occurrences of a particular allele in a population. (1)
  - c. The process by which organisms become better adapted to their environment due to favourable characteristics, which allow them to be more likely to survive and reproduce. (1)
4. [10]
  - Sickle-cell anaemia is caused by inheriting two alleles of a mutant gene, (1)
    - that results in abnormal haemoglobin / mutated form of haemoglobin / distortion of red blood cells / reduced oxygen-carrying capacity of red blood cells. (1)
    - It results in early death in those who have both alleles. (1)
    - Individuals with one allele has partial sickle-cell anaemia, but generally, survive. (1)
    - In countries where malaria is prevalent, e.g. Western Africa. (1)
    - there is higher incidence of the sickle-cell trait. (1)

- compared to countries like Australia. (1)
  - Those who have the sickle-cell trait are less susceptible to malaria than those who don't have the trait. (AW) (1)
  - This provides a selective advantage / survival advantage in Western African countries. (1)
  - This is not the case in Australia, so the sickle-cell trait is not selected for. (1)
5. [8]
- Mutations to genes results in variation within the gene pool. (1)
  - Some of these mutations provide a selective advantage. (1)
  - These is a struggle for existence / there is limited resources. (1)
  - So those individuals with the advantageous alleles are more likely to survive to reproduce. (1)
  - The advantageous alleles are passed onto the next generation. (1)
  - Resulting in an increase in these advantageous alleles / genes over time or over several generations. (1)
  - In this way, new groups evolve. (1)
  - by the process of natural selection. (1)

| Absolute                                    | Relative                                    |
|---------------------------------------------|---------------------------------------------|
| Works best on igneous and metamorphic rock. | Works best on sedimentary rock.             |
| Provides the order of formation of remains. | Provides the order of formation of remains. |
| Provides the specific age of remains.       | Provides a rough age of remains.            |

- Any seven comparisons for one mark each. No ½ marks. [7]
5. Bioinformatics is the science of collecting, analysing and understanding complex biological data using computational techniques to analyse the information. (1)  
It can be used to compare the genomes/ genes of different organisms to see how many similarities/differences they have. (1)  
Using this technique it can be seen how closely related different species/organisms are. (1) [3]
- 6.
- a. Polymerase chain reaction (A: PCR) [1]
  - b. Gel electrophoresis. [1]
  - c. Comparative studies are conducted to study the relatedness of species. (1)  
One from:  
The similarities and differences can help us to determine when the common ancestor was. (1) (AW)  
The differences can help understand how modern humans and Neanderthals evolved. (1) (AW) [2]
  - d. Finding enough DNA to extract. (1)  
Finding DNA that hasn't been destroyed due to decay. (1)  
Avoiding contamination with modern human DNA (as they are closely related). (1)  
Any two for one mark each. [2]
7. Embryology: The study of the early development of an organism. (1)  
Comparative studies of homologous structures: Comparing the structures of organisms that have similar anatomy, but carry out different functions. (1) [2]

**Chapter 8  
Evidence for Evolution (Unit 4)**

1. Any three of the following for one mark each: quick burial, hard bone parts, lack of decay organisms, limited disruption to the soil, no oxygen, alkaline conditions (A: acid conditions if it is wet and no oxygen is present). (Only accept first three answers. A fourth or more answer would be ignored by an examiner). [3]
2. Vestigial structure is reduced in size and has appears to have no function. (1)  
Appendix contain some bacteria which may assist digestion, so this means the appendix has a function. (1) [2]
3. Carbon-dating is only effective up to 60,000 years. (1)  
Homo erectus is 60,000 years old BP. (1) [2]
4. The best way to answer this question is as a table so that contrasts and similarities can be seen:

| Absolute                                                                                                | Relative                                                                                                         |
|---------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------|
| Determines the age of a specimen using radiometric techniques / Determines the exact age of a specimen. | Determines the position of a specimen using stratigraphy / Determines the age of specimen in relation to others. |
| Is quantitative.                                                                                        | Is qualitative.                                                                                                  |
| Is more specific.                                                                                       | Is less specific.                                                                                                |
| Relatively expensive.                                                                                   | Relatively cheap.                                                                                                |
| Relatively time-consuming.                                                                              | Relatively quicker process.                                                                                      |

**Chapter 9  
Trends and Culture in Hominid Evolution (Unit 4)**

1. [6]
  - Foramen magnum (1): Has moved more centrally so it can balance on top of vertebral column. (1)
  - S-shape / curvature of spine (1): S-shape contributes to upright stance (1)
  - Reduced prognathism (1): Doesn't stick / jut out, allowing skull to balance on vertebral column (1)

2.

| Australopithecines                                                                       | Homo erectus                                                                    |
|------------------------------------------------------------------------------------------|---------------------------------------------------------------------------------|
| • Use of pebble tools. (1)                                                               | • Use of Acheulian tools / tools modified for purpose. (1)                      |
| • Made of stone. (1)                                                                     | • Made of stone and bone. (1)                                                   |
| • Examples: chopper, scraper, flakes, chisels<br>(Any suitable example for one mark) (1) | • Examples: hand-axes, spears, hooks<br>(Any suitable example for one mark) (1) |

*This could be written as comparative dot points, but it cannot be a paragraph on the australopithecines followed by a paragraph on Homo erectus, because this is not comparative.*

[6]

3.

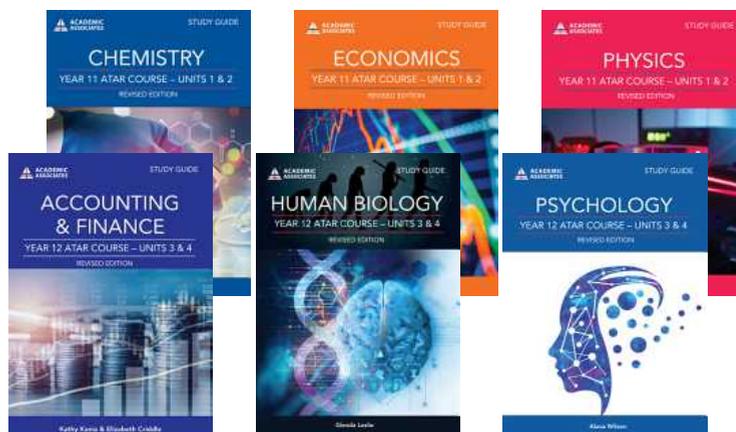
- Potassium-argon dating / K-Ar dating. (1)
  - Potassium-40 has a half-life of 1.25-1.3 billion years. (1)
  - By comparing the amount of potassium-40 relative to argon-40 (1)
  - determines the age (of the volcanic rock). (1)
  - Needs to be over 100, 000 years (and less than 200, 000 years). (1)
4. [5]
- By making / using endocasts. (1)
  - Increased size (of the frontal lobe) (1)
  - from 430 cm<sup>3</sup> (1)
  - to 1350 cm<sup>3</sup>. (1)
  - Increased convolutions. (1)

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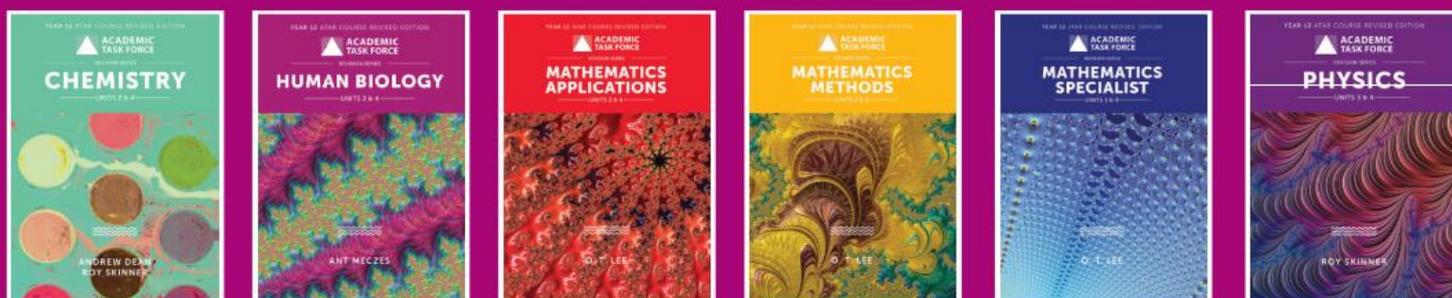


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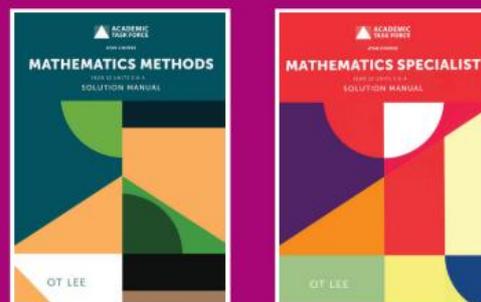
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