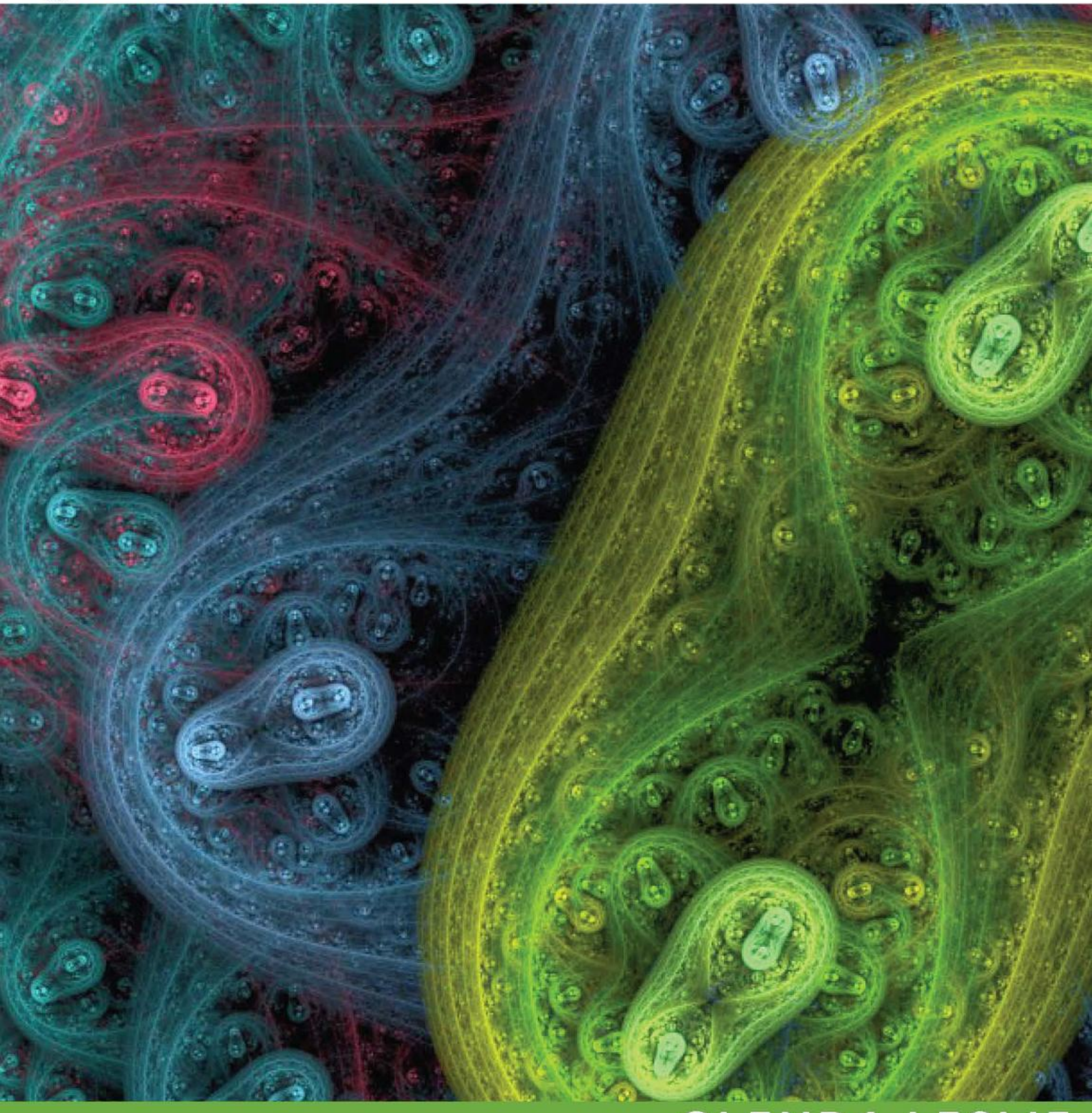
ACADEMIC TASK FORCE

HUMAN BIOLOGY

YEAR 11 ATAR COURSE UNITS 1 & 2

REVISION SERIES - FIRST EDITION



GLENDA LESLIE

ACADEMIC
TASK FORCE

REVISION SERIES

HUMAN BIOLOGY

YEAR 11 ATAR COURSE

Glenda Leslie

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About the Author
Glenda Leslie was Chief Examiner for the Human Biology TEE paper and as exam reviewer for the ATAR Human Biology examination. She worked at the Curriculum Council (SCSA), as project officer for the development of the Year 11 and 12 courses for Human Biological Science. She has been involved in the development of the Australian Curriculum for Biology as a representative from Western Australia on the ACARA advisory panel. Glenda was an inaugural member of the School Curriculum and Standards Authority Curriculum Advisory Committee for Human Biology for over ten years, guiding the changes in content and assessments requirements. Glenda has also produced workbooks for Science Teachers Association of WA and a study guide for Academic Associates for ATAR students studying Human Biology.

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INTRODUCTION

The Academic Task Force Revision Series is a revision resource for students to use alongside class textbooks and lesson notes. This Revision Series book, enables students to review and improve their understanding of concepts for Human Biology Year 11, Units 1 & 2 in preparation for tests and exams. Chapters include a syllabus checklist against each learning stage (Discover = core theory and initial syllabus checkpoint teaching, Revise = revision of the syllabus checkpoint, Enrich = further investigation and understanding of syllabus checkpoint, Exam Prep = ability to answer and demonstrate the syllabus checkpoint). Each chapter has revision notes on concepts and questions to revise content. Answers are provided at the back of the book.

This revision book uses figures, tables and images that cover the content. Students should observe and annotate these carefully and follow the sequence of processes shown to relate the notes to the images. Students are encouraged to add extra information or arrange the content in other forms that will help them understand the concepts. Diagrams and flow charts are excellent ways to develop a student's understanding of the body systems and their interactions.

The chapters include tasks. At these points, students are asked questions based on the content. To answer the task question, students may check the notes provided, review their class notes, discuss with others and use other resources to write a considered response. Most chapters conclude with revision questions. Revision questions should be undertaken once the revision notes are understood and tasks are completed. Attempt revision questions multiple times to prepare for your examinations as questions are styled in the format and difficulty you would expect in an exam covering the examinable content.

The ATAR Human Biology exam design brief is outlined below.

Section one: Multiple choice

> Questions can require the candidate to refer to the stimulus material that can include text, diagrams, second-hand data and/or graphs.

Section Two: Short answer

> Each question is divided into parts. Typically, the parts within a question are of increasing difficulty.
>
> Questions can require the candidate to refer to the stimulus material that can include text, diagrams, second-hand data and/or recent research material.

Section Three: Extended answer

> Responses could include clearly labelled diagrams with explanatory notes, lists of points with linking sentences, clearly labelled tables and graphs and annotated flow diagrams with introductory notes.

This book will help students organise their knowledge in ways that can be adapted to answering questions using stimulus material in the form of diagrams, tables, graphs and text. Answers can also be in these forms so students need to know how to read and construct each form.

This book will also help students organise their learning to improve their understanding of their own body as well as those of their relatives and friends, whether they be young or old, what stages of development they have been through and what is expected next. The course content is finished with the chapter on lifestyle choices: decisions that have short and long-term consequences for each person, their family and their possible offspring. Choose wisely!

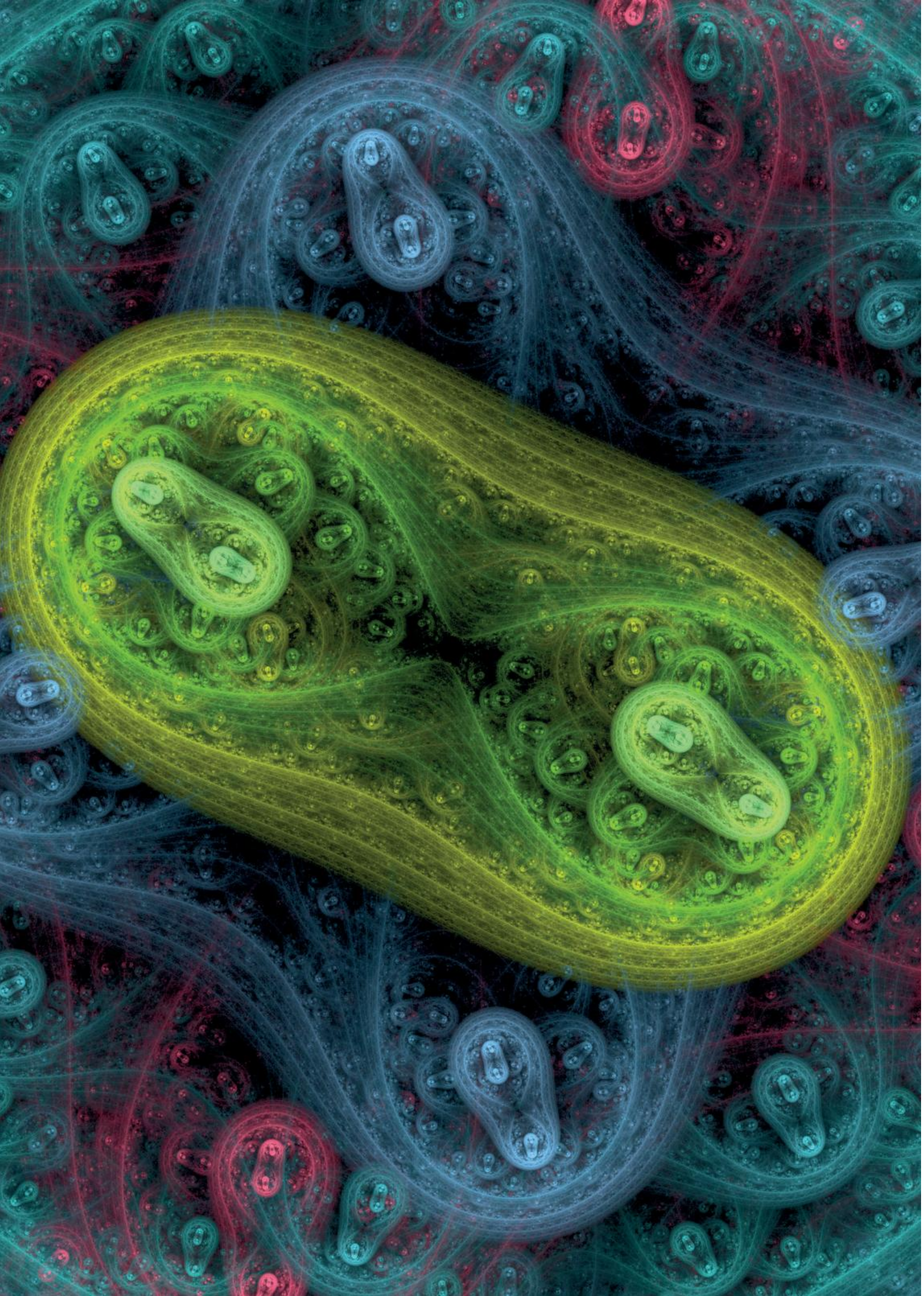
May you live a long and healthy life, including being successful in any human biology exam.

Cheers,

Glenda Leslie

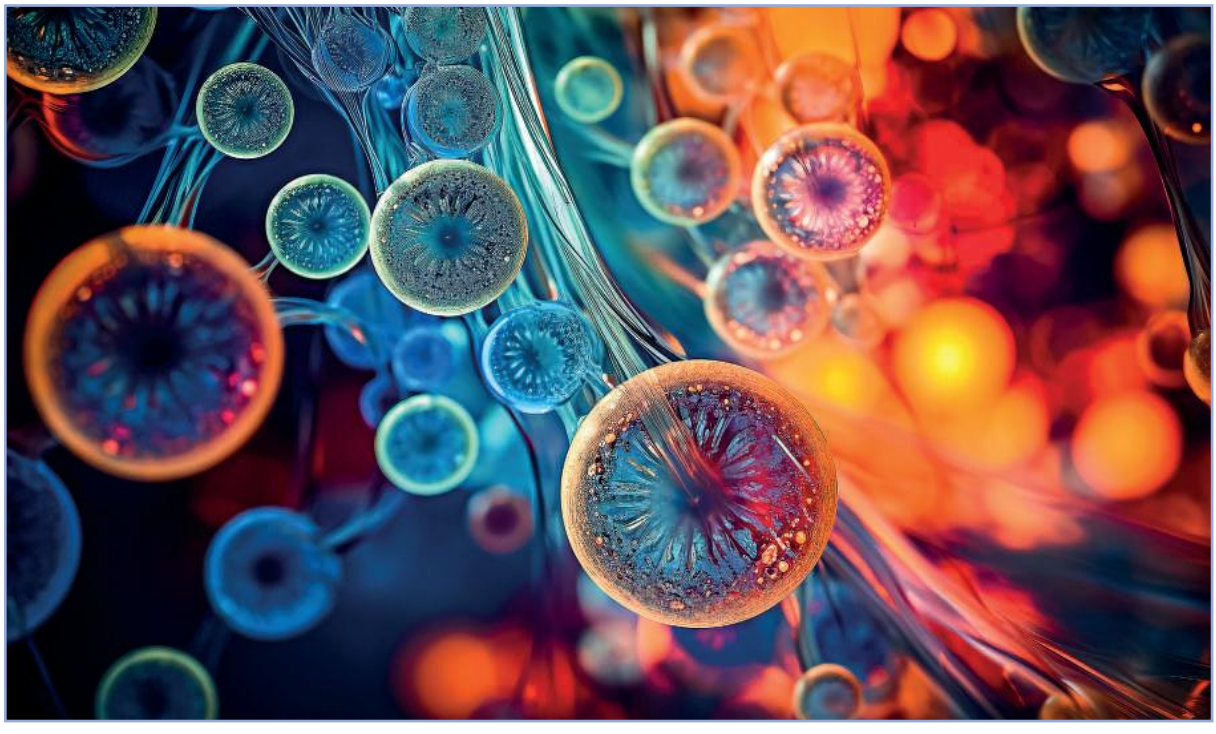
CONTENTS

UNIT TWO REPRODUCTION AND INHERITANCE



CELLS AND BODY HIERARCHY

1



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The human body is comprised of cells, tissues and organs within complex systems that work together to maintain life.				

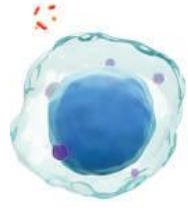
All living things are made up of cells and the products of cells.

All living cells need to carry out the following functions to survive.

Movement: can move the whole body or its parts independent of external influences; can be voluntary or involuntary

Growth: the body becomes larger and more complex as it changes from the embryo to the adult organism; materials are incorporated into existing body structures

Cellular structure: all living things are made up of cells and the products of cells



Reproduction: adults produce young that are similar to themselves

Respiration: the release of energy from food for metabolic uses; can be aerobic (with oxygen) or anaerobic (without oxygen)

Excretion: waste products of metabolism or excess materials are removed from the body the form of solids (faeces), liquids (urine) or gases (carbon dioxide)

Sensitivity: can detect changes in the internal or external environment and respond to these changes

Nutrition: intake of food that provides energy for metabolism and building materials for growth

Figure 1.1: Characteristics of living things

Table 1.1: Comparing living and non-living things

Feature	Living	Non-Living
Organisation	Highly organised	Very little or no organisation
Responsiveness	Sense and react to external stimuli	Do not react to stimuli
Homeostasis	Maintain a stable internal environment	Do not maintain a stable internal environment
Metabolism	Use of energy from the breakdown of materials to build required materials	No energy use
Growth	Undergo regulated growth through internal processes	Grow only if materials are added to the external surface e.g. crystals
Adaptation	Groups of living things can change to suit their environment over time	Do not form groups that produce changes over time
Survival	Require inputs and outputs to support internal processes	No internal processes present; no inputs or outputs
Lifespan	Have a life span, then die	Life span not applicable
Reproduction	Ability to produce others similar to themselves through internal processes	Do not reproduce

Task 1.1: Is a car a living thing? Explain your reasoning.

BODY HIERARCHY OF ORGANISATION

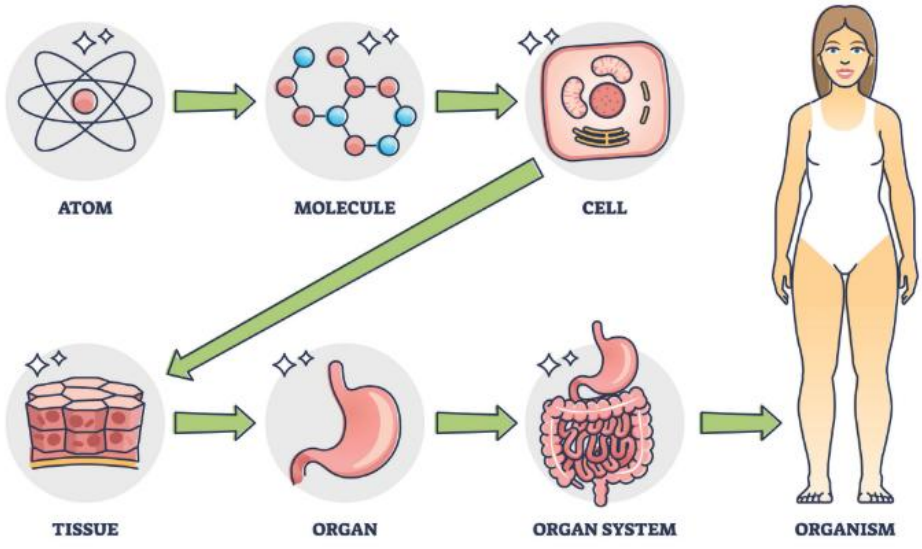


Figure 1.2: Organisation in organisms

All matter is made up of atoms and can combine to form molecules.

- **Cell**: Basic unit of structure and function of all living things.
- **Tissue**: Group of cells of the same kind.
- **Organ**: Structure composed of one or more types of tissues working together to perform a specific function.
- **Organ system**: Group of organs that work together to perform a certain function related to the features of living things.
- **Organism**: Individual living thing that may be made up of one or more organ systems.

BODY SYSTEMS

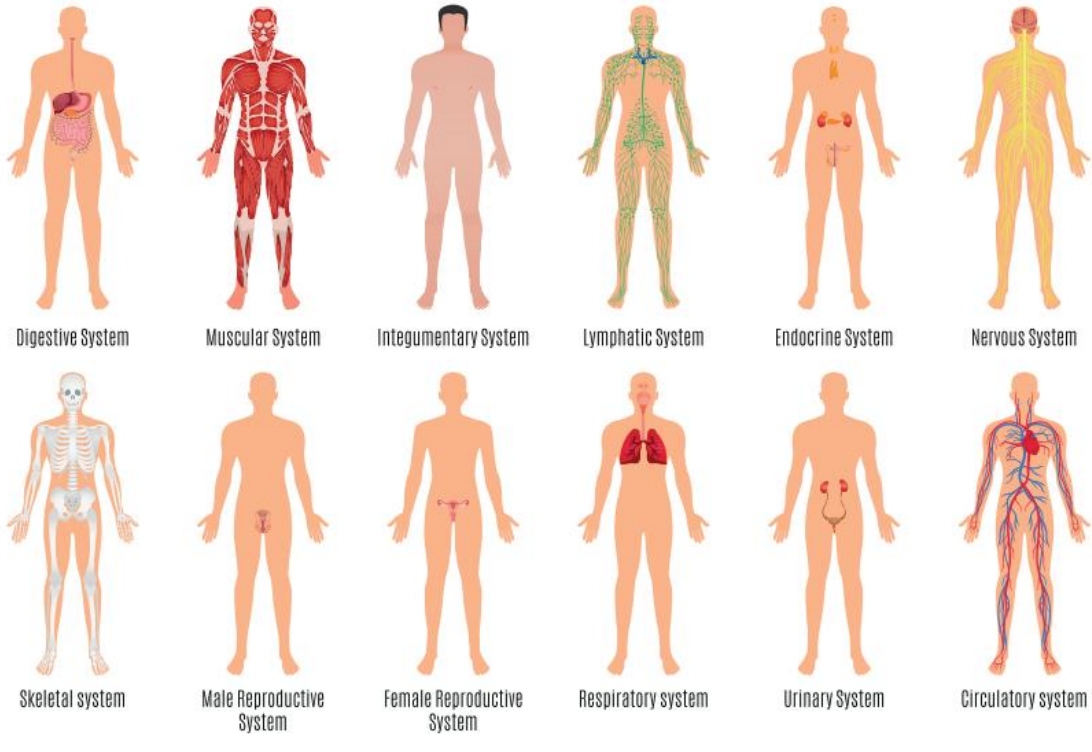


Figure 1.3: Systems of the human body

System: A group of organs that work together to perform one or more functions in the body.

Task 1.2: Complete the following table by adding the organs associated with each system.

Systems	Function	Organs
Musculoskeletal system includes muscles and skeleton	Mechanical support, protection, posture and locomotion Production of blood cells and storage of minerals and fats	
Circulatory system includes cardiovascular, lymphatic and immune systems	Transportation of gases, nutrients and hormones throughout the body and elimination of cellular metabolic waste Draining of excess tissue fluid, immune defence of the body	
Respiratory system	Exchange of oxygen and carbon-dioxide between the body and air, phonation	
Nervous system	Regulates behaviour, helps maintain homeostasis and controls sensory and motor functions	
Endocrine system	Production of hormones to regulate a wide variety of bodily functions (e.g. menstrual cycle, blood sugar levels, body temperature)	
Digestive system	Mechanical and chemical breakdown of food to simple nutrients for absorption into the body cells	
Urinary or excretory system	Filtration of blood and elimination of toxic and excess compounds and waste, regulates concentration of body fluids	
Reproductive system	Production of reproductive cells and support of the developing offspring (females)	
Integumentary system	Physical protection of the body surface, sensory reception, vitamin synthesis; preventing fluid loss and helps regulate body temperature	

Task 1.3: For the organs shown in Figure 1.4, state the system to which each belongs.

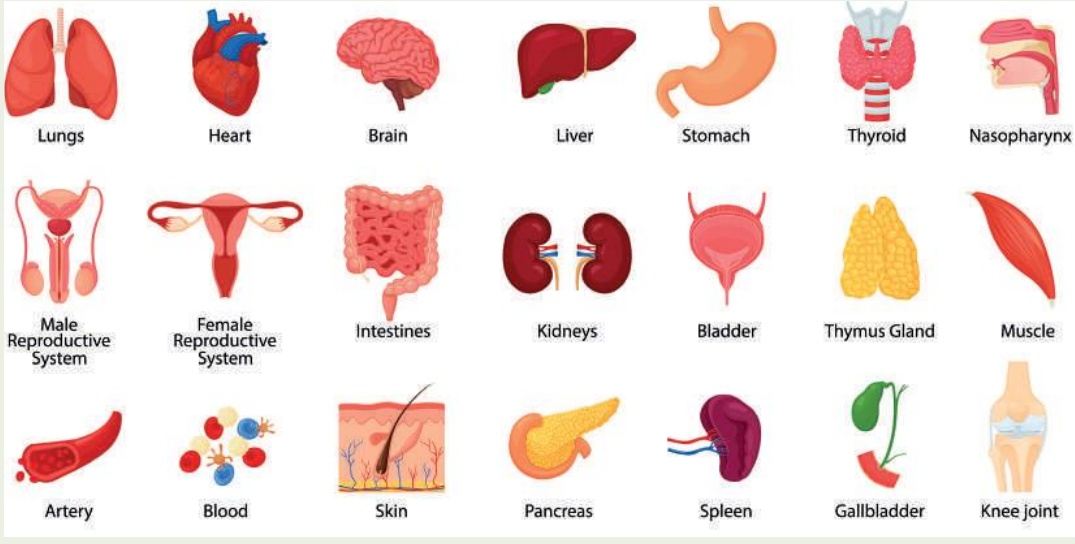


Figure 1.4: Organs of different body systems

SYSTEMS SUPPORTING CELLS

The only reason organs exist is to support the functions of all individual body cells.

Body cells are specialised in some functions to support other cells. Cells need to work together to bring about the movement of the whole body, protect the body against disease, respond to the environment, grow and reproduce.

Not all body cells have direct access to the external environment to exchange materials. Body systems specialise in these exchanges e.g. circulatory system, digestive system, respiratory system and excretory system.

The whole body requires a control system to produce co-ordinated responses to the external environment and maintain a constant internal environment. The nervous and endocrine systems are responsible for this.

Table 1.2: Systems supporting cells

Cells	Characteristics of Living Things	Systems
Internal cytoplasmic movements Whole cell movements	Movement	Musculo-skeletal systems
Cellular respiration (not to be confused with breathing or gas exchange)	Respiration	Respiratory system
Cell membrane (receptors) Nucleus (DNA) Cellular organelles (chemical reactions)	Stimulus response	Nervous system Endocrine system
Mitosis	Growth	All systems have finite and sequenced growth patterns
Mitosis / Meiosis	Reproduction	Reproductive system
Removal of waste and excess materials across cell membrane	Excretion	Excretory system – kidneys Integumentary system – skin exchange surfaces in digestive system and respiratory system
Intake of nutrients	Nutrition/ingestion	Digestive system
No specialised system for transport at cellular level		Circulatory system

Question 1

Name the systems that work together to allow for the exchange of gases between cells and the external environment. (2 marks)

Question 2

Name the system(s) that:

(a) are involved in walking. (3 marks)

(b) is the main system difference between males and females. (1 mark)

(c) controls long term actions such as growth. (1 mark)

(d) share structures. (3 marks)

(e) individuals can survive without. (1 mark)

Question 3

Explain why hair is non-living, but bone is living. (2 marks)

Question 4

Fingernails and toenails are made of dead cells. How can the accumulation of dead cells (your nails) be used to indicate the life process of 'growth'? (3 marks)

Question 5

How can you tell if a person is unconscious or dead? (7 marks)

Question 6

What is the final and irreversible sign of death? (1 mark)

Question 7

Which systems have openings to the outside of the body? (4 marks)

Question 8

State the general difference between tissues and organs. (4 marks)

Question 9

Explain what changes would occur in the body if erythrocytes (red blood cells) were removed as is the case in anaemia. (6 marks)

Question 10

Explain the impacts to digestion if the cells lining the digestive tract did not produce chemicals such as enzymes and acid. (4 marks)

Question 11

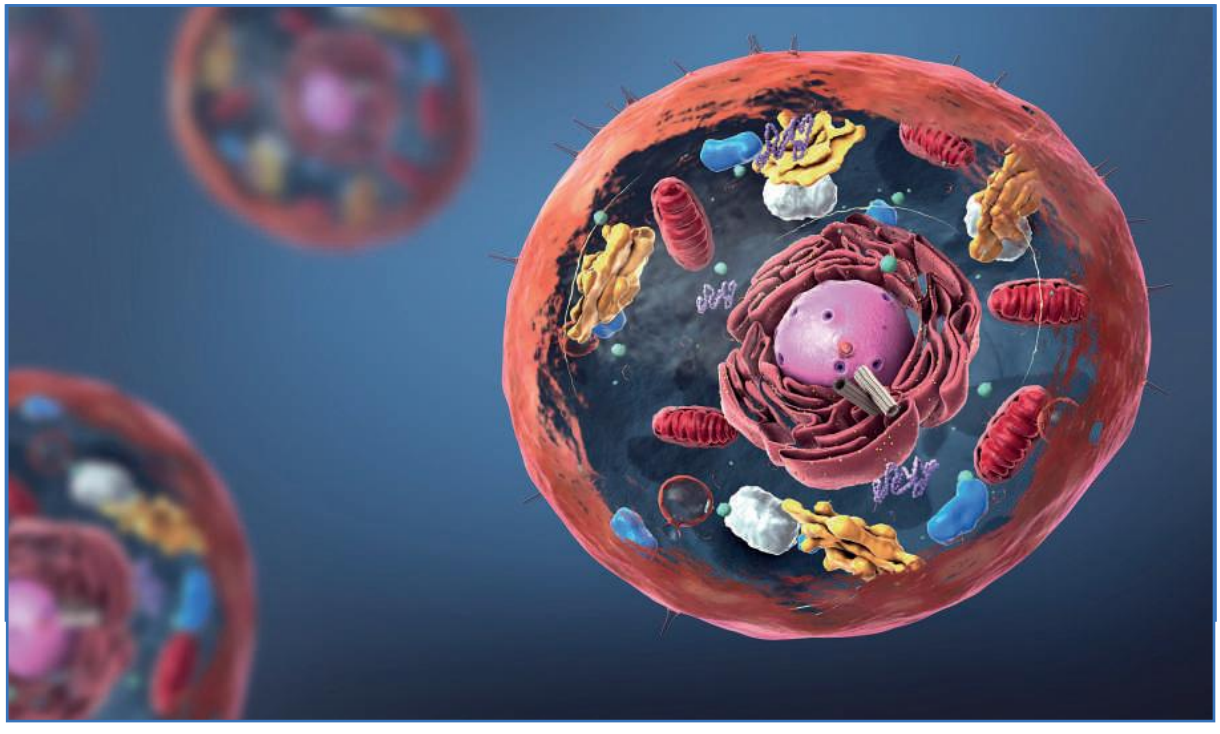
If the digestive system of a person fails, describe how medical intervention can still supply nutrients to cells. (2 marks)

Question 12

Venom from the blue-ringed octopus stops muscle contraction. Describe why people die as a result of being bitten by the blue-ringed octopus. (3 marks)

CELLULAR STRUCTURE

2



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Cell organelles maintain life processes and require the input of materials and the removal of wastes to support efficient functioning of the cell.				

Cells are the basic unit of living things.

Cells have specialised structures that carry out functions **efficiently** to supply a cell with its requirements at **sufficient** rates to maintain its overall role in the body.

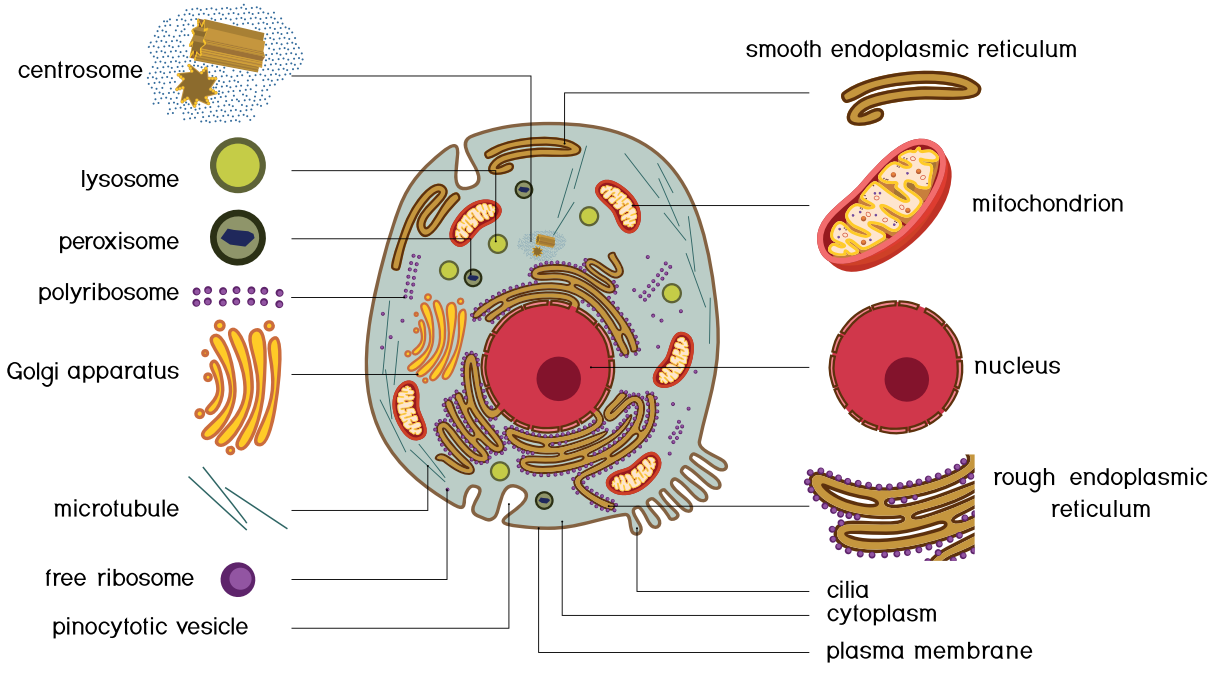


Figure 2.1: Generalised cell structure

No human body cells look like the figure of the generalised cell above.

Task 2.1: Explain why a generalised cell is used for describing the contents of body cells.

Table 2.1: Cellular components

Organelle	Components	Function
Nucleus	Chromosomes Nucleolus	Contains genetic materials; assembly of ribosomes
Ribosomes	RNA and proteins	Site of protein synthesis
Mitochondria	Enzymes attached to inner membrane	Site of aerobic respiration
Cell membrane	Phospholipid bilayer with protein channels and receptor proteins	Maintains the cellular environment by controlling the movement of materials into and out of the cell; receptor for information on external environment
Golgi body	Stack of flattened discs of membranes	Protein processing
Endoplasmic reticulum	Flattened branching sacs with or without ribosomes attached	Protein processing and lipid synthesis
Lysosomes	Double membrane vesicle which contain catalase enzymes	Breakdown of excess or damaged organelles and materials entering the cell by endocytosis
Cytoskeleton	Filaments and microtubules	Structural support and shape of the cell; movement of materials within the cell and the whole cell
Vesicles	Fluid filled phospholipid bilayer capsule	Storage of materials for secretion
Cilia	Hairlike extension of the cell membrane	To move the cell or materials across the cell
Centriole	Microtubules	Involved in the organisation of the mitotic spindle

Question 1

Figure 2.2 shows a 3D version of the cell in Figure 2.1. Use the information from Figure 2.1 to label the structures shown in Figure 2.2. (14 marks)

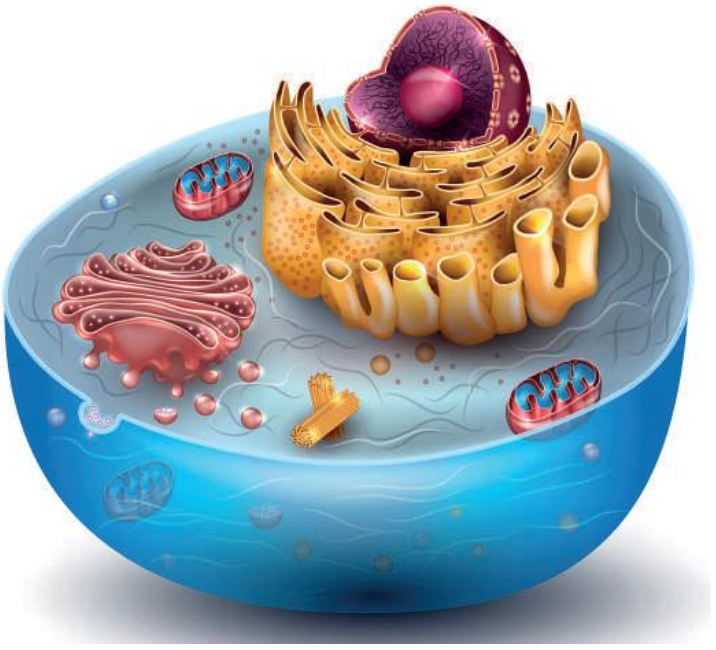


Figure 2.2: Generalised human cell

Question 2

Answer the questions with reference to Figure 2.3.

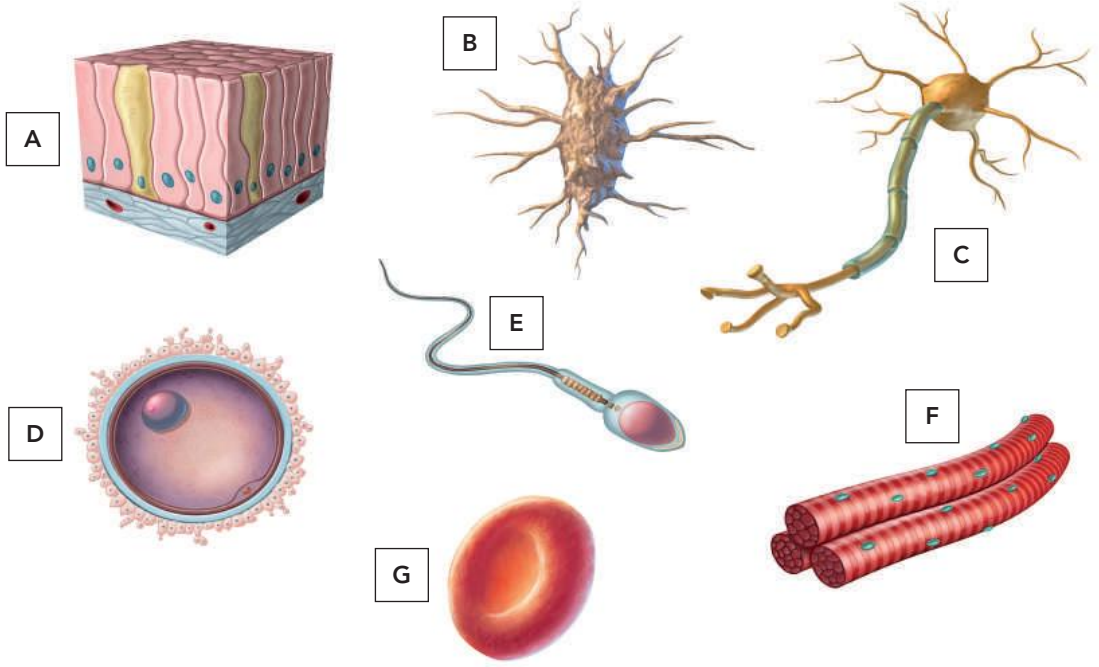


Figure 2.3: Variety of body cells and their organelles

(a) Which of A–G show single cells? (3 marks)

(b) For A–G that are not single cells, circle a single cell. (3 marks)

(c) For A–G, label the nuclei shown. (4 marks)

(d) What is the main difference between E and the rest of the cells in the diagrams? (1 mark)

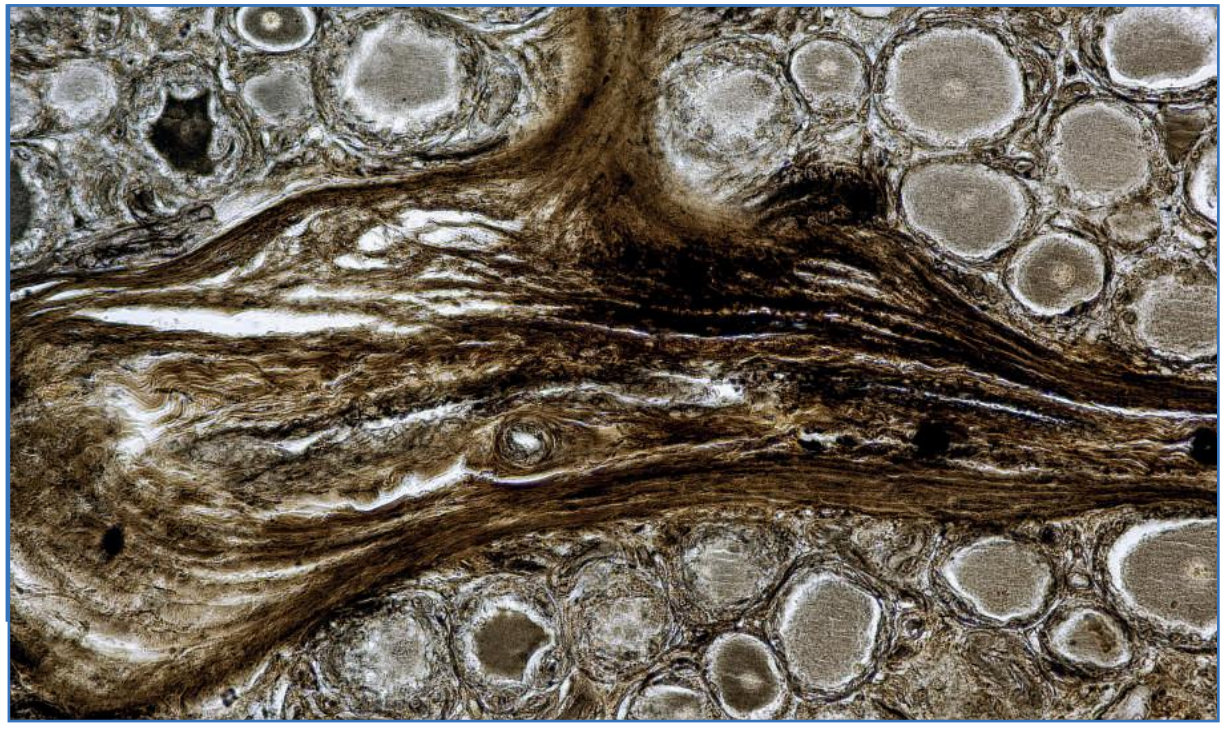
(e) State the common feature of cells in diagrams B and C. (1 mark)

(f) Cells in diagrams E and F have a greater number of mitochondria than the generalised cell. Suggest a reason for this. (2 marks)

(g) The cell shown in G does not have a nucleus. What cellular activities would not occur in this cell? (3 marks)

(h) The yellow cells in A produce secretions. What organelles would these cells have in greater numbers than a generalised cell? (4 marks)

3 TISSUE TYPES AND STEM CELLS



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The various tissues of the human body perform specific functions and can be categorised into four basic tissue types: epithelial, connective, muscular and nervous.				

Body organisational hierarchy:

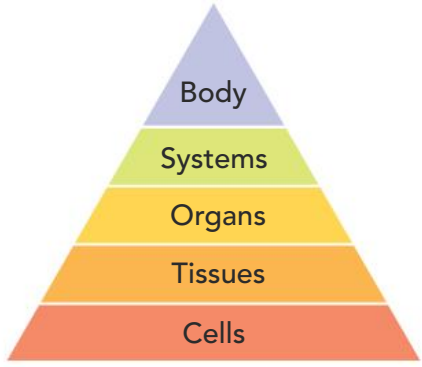


Figure 3.1: Body Organisation Hierarchy

TISSUE TYPES

Similar cells with similar functions in the same location form a **tissue**.

The cells of tissues have **similar cellular contents, functions and shapes**.

They can be classified according to the differences in these factors.

Cells with more mitochondria will most likely carry out respiration at a greater rate than other cells. These will be found in tissues that require a lot of energy either to move or to produce materials to secrete.

Cells with long extensions most likely need to be able to connect to other cells at a distance away e.g. neurones or osteocytes.

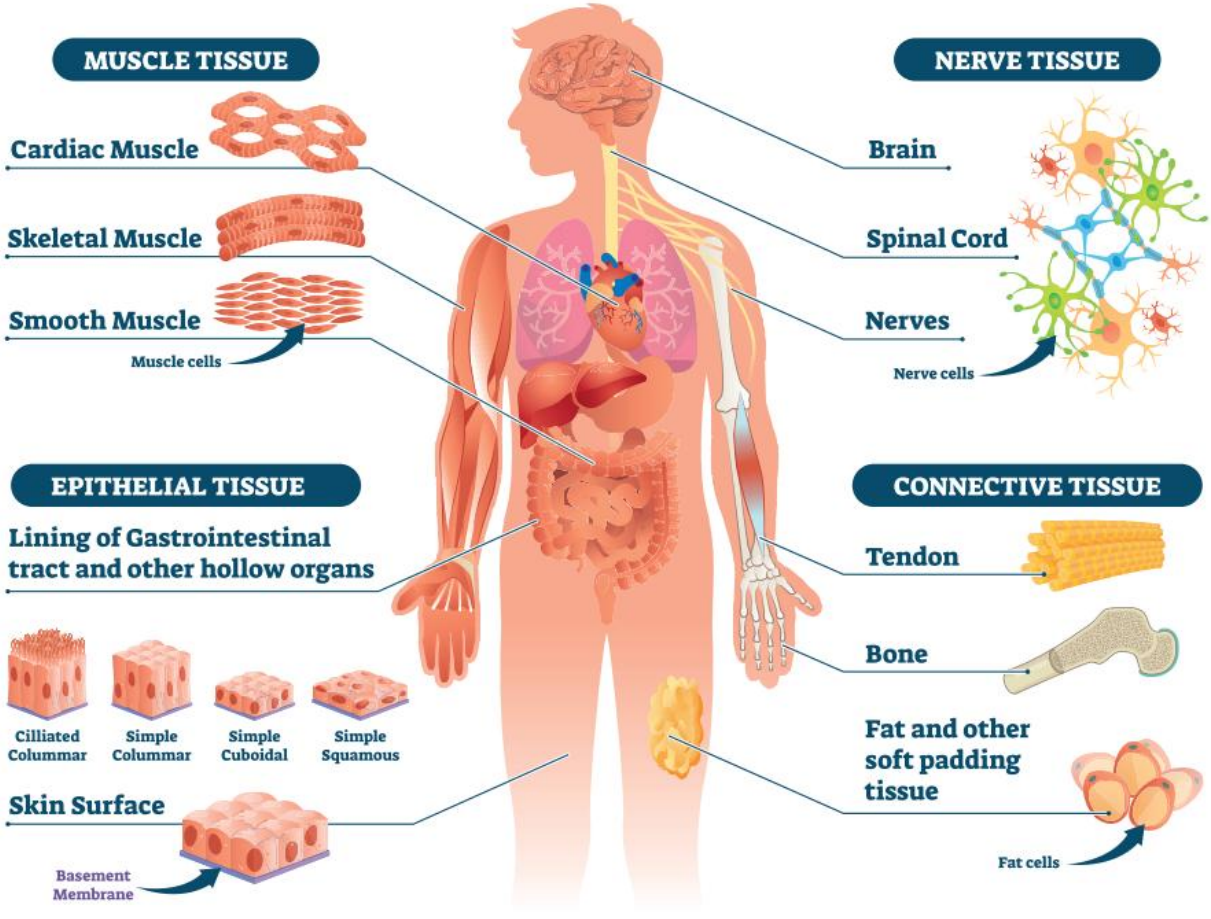


Figure 3.2: Classification of tissues and their sub-groups

Table 3.1: Classification of tissues according to their structure and function

Category	Types	Function	Locations
Muscle tissue • Contraction – ability to shorten in length	Skeletal/voluntary	Movement at joints – controlled contractions	Attached to the bones of the skeleton
	Cardiac	Pumping of blood	Heart
	Smooth/involuntary	Involuntary movements: contraction of blood vessels and airways, peristalsis – waves of contractions	Surrounding the blood vessels, the digestive tract and the larger respiratory tubes
Epithelial tissue • Covering tissue	Simple squamous	Allows for diffusion of materials	Alveoli of lungs
	Simple cuboidal	Involved in secreting (producing and releasing) and absorbing substances in various glands and ducts	Lining the ducts of kidney, pancreas, ovaries, salivary glands
	Simple columnar	Produce secretions especially mucus for surfaces	Digestive tract and female reproductive system
	Ciliated columnar	Movement of mucus across surfaces by waving action of cilia	Lining the trachea and bronchi of the respiratory tract, and the small intestine
	Stratified	Includes dead cells and provides waterproofing for skin	Skin Around ducts of mammary glands
Nerve tissue • Ability to conduct electro-chemical impulses • Cells are called neurones	Grey matter – contain neurones without myelin sheaths	Transmission of nerve impulses	Centre of spinal cord Outside covering the brain
	White matter – contains neurones with myelin sheaths	Schwann cells produce myelin to allow faster transmission of nerve impulses	Outside area of the spinal cord Internal parts of the brain
Connective tissue • Joining tissues	Bone – cells are called osteocytes	Provides inflexible support for muscles	The main tissue of the skeleton
	Cartilage – cells are called chondrocytes	Provides flexible support for body parts such as nose and ears	End of long bones, in other joints, end of the nose, ear pinna
	Blood – cells include, erythrocytes and leucocytes; platelets are cell fragments	Transport gases, nutrients, hormones, distributes body heat, protection against pathogens	In blood vessels of the circulatory system
	Loose connective tissue	Connects epithelial tissue to basement membranes	Skin, underneath epithelial tissues
	Adipose tissue	Protection for organs, storage of lipids	Fat, padding around eye sockets and joints
	Dense connective tissue	Protection against trauma	Layer around internal organs
	Ligaments	Join bones to bones	All synovial joints
	Tendons	Join muscles to bones	All muscle attachments to bones
Reproductive tissue	Female Male	Ability to produce Gametes by meiosis	Ovaries Testes

Different tissues types work together to form organs. Each tissue type has its own function to work with others to bring about the overall function of the organ.

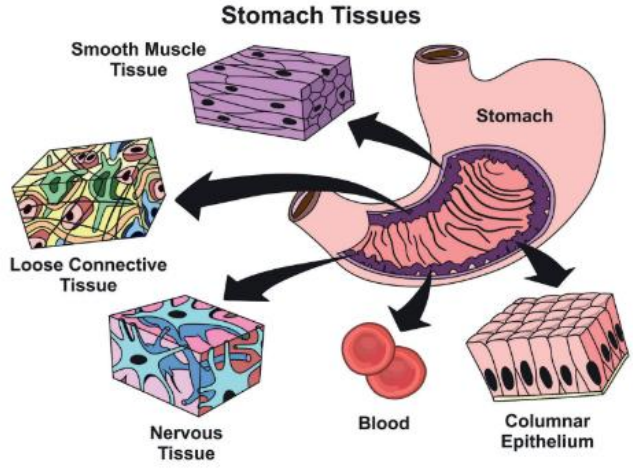


Figure 3.3: Tissues that make up the stomach

Task 3.1: State the function of each of the tissue types shown in Figure 3.2 for the proper functioning of the stomach.

EPITHELIAL TISSUE

Epithelial tissue is formed from a tightly fitted continuous layer of cells. One surface of the epithelial tissue is exposed to either the external environment or a body fluid. The other surface is attached to tissue by a membrane, which consists of fibres and polysaccharides secreted by epithelial cells.

There is little intercellular material present between cells.

Epithelial cells form membranes. The epithelial membrane consists of a layer of epithelial tissue and has underlying connective tissue.

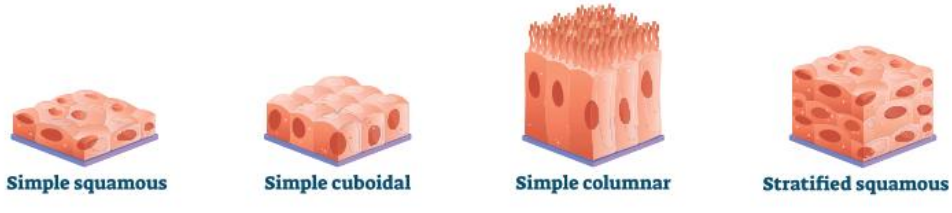


Figure 3.4: Epithelial tissue types

Task 3.2: Complete the boxes beneath the types of epithelial tissues in Figure 3.3 indicating where they can be found in the body.

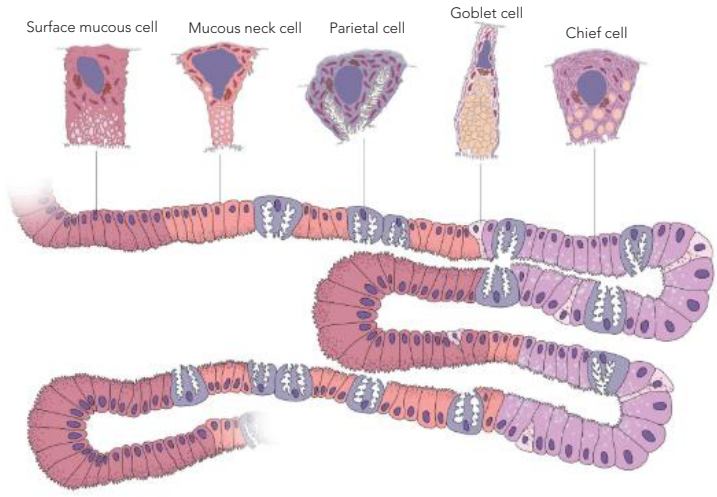


Figure 3.5: Types of cells found in the lining of a gastric pit in the stomach

The epithelial tissue lining the gastric pits of the stomach are made up of a variety of cells, each with specific functions and cellular processes and therefore contents.

- Parietal cells secrete hydrochloric acid.
- Chief cells secrete pepsinogen (a precursor to pepsin – the enzyme that digests proteins).
- The surface mucous cells, mucous neck cells and goblet cells all produce and secrete mucus onto the surface of lining the gastric pit and the stomach.

But all of them have at least **some part of their surface that is free of contact with other cells** i.e. facing the space in the gastric pit. The free surface is the common **feature of epithelial tissues**.

MUSCLE TISSUE

Muscle tissue has the **ability to contract** causing movement.

There are **three** different types of muscles for three different types of movement.

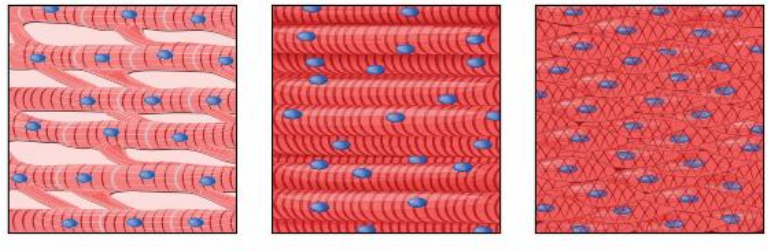


Figure 3.6: Muscle tissue types

Table 3.2: Features of different muscle types

Cardiac muscle	Skeletal muscle/ voluntary muscle	Smooth muscle/ involuntary muscle
Produces rhythmic muscle contractions	Produces discrete muscle contraction	Contracts slowly and automatically
Found only in the heart	Found attached to bones	Found in the walls of passageways such as the digestive tract, blood vessels and uterus
Striated	Striated	Non-straited
Involuntary control	Voluntary control	Involuntary control
Has cross branches connecting muscle cells	Individual muscles cells in bundles	Spindle-shaped cells

CONNECTIVE TISSUE

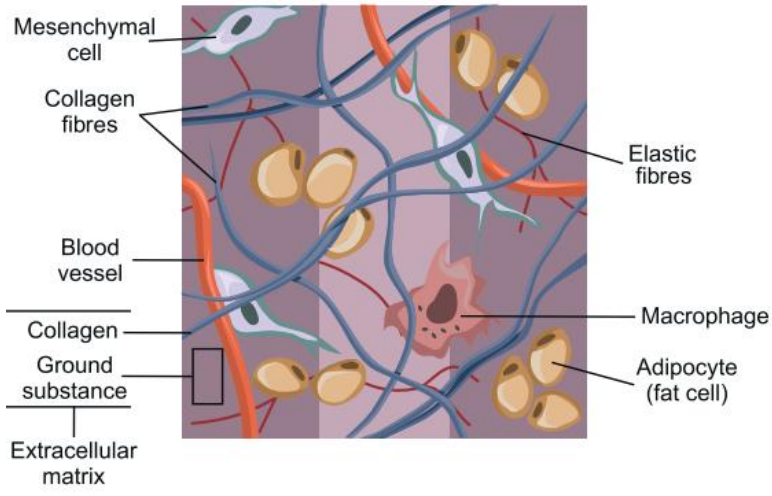
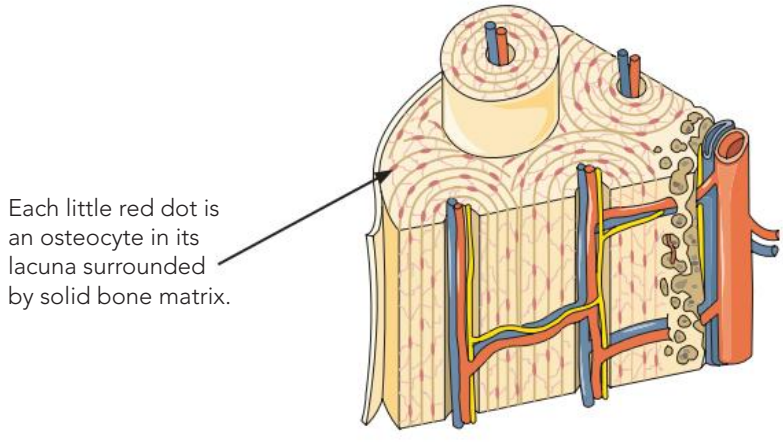


Figure 3.7: Generalised connective tissue components

BONE TISSUE



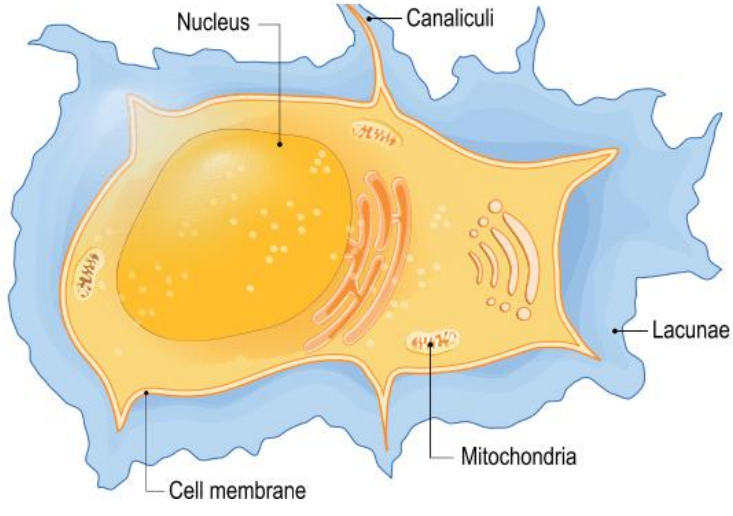


Figure 3.8: Bone tissue and location of osteocytes

CARTILAGE TISSUE TYPES

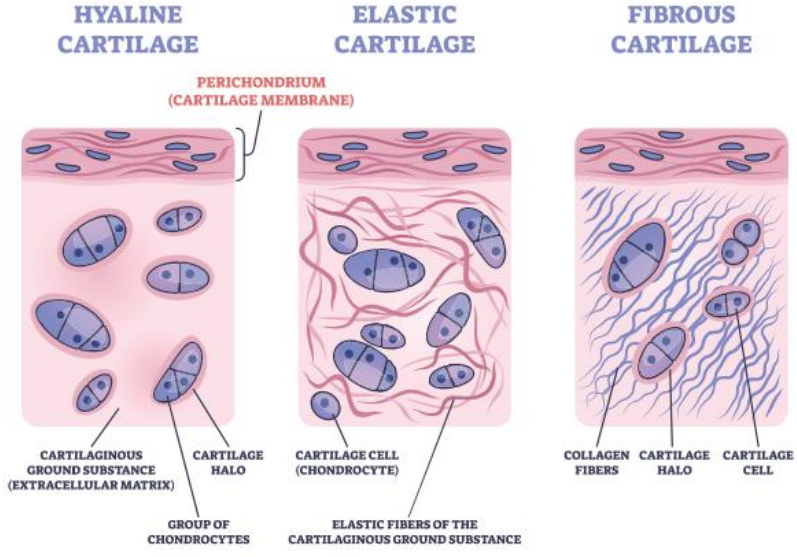


Figure 3.9: Types of cartilage

ADIPOSE TISSUE

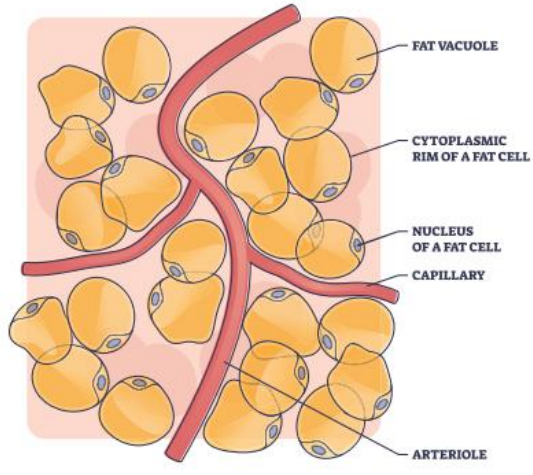


Figure 3.10: Adipose tissue

BLOOD

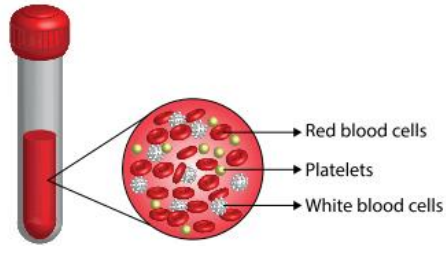


Figure 3.11: Blood cellular contents

Connective tissue is made up of cells set in a matrix, which may contain fibres such as collagen, elastic fibres or reticular fibres.

The main functions of connective tissue are:

- binding and supporting – ligaments, tendons and bones
- insulating organs and energy storage – adipose tissue
- transport of materials - blood and lymph
- protecting organs – loose connective tissue and adipose tissue.

Types of connective tissue

Loose connective tissue:

- where support and elasticity is required, such as joining skin to muscles, between organs as filling and shock absorbers and reservoirs of fat, fluids and salts.

Dense connective tissue:

- where support is required and to transmit mechanical forces e.g. tendons and ligaments.

Specialised connective tissue:

- help maintain posture and support internal organs e.g. bone and cartilage
- fluid connective tissues such as blood and lymph for transport of materials around the body.

NERVOUS TISSUE

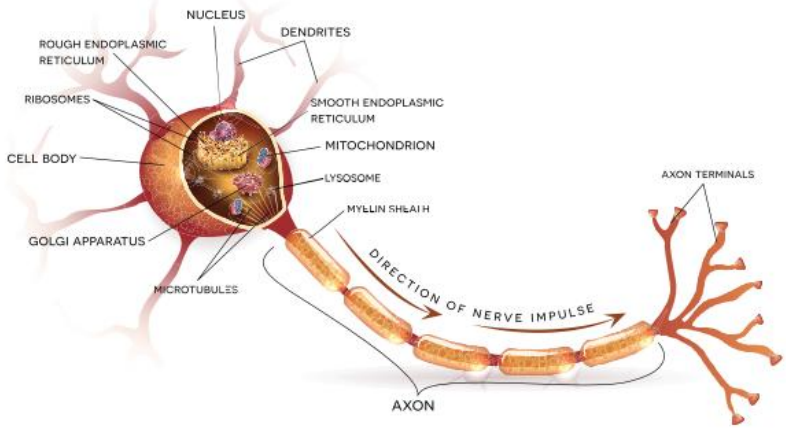


Figure 3.12: Neurone structure

Nerve tissue is made up of neurones which have long cellular extensions that make connections with other neurones or tissues such as muscles or glands.

– **Axons** transmit nerve impulses away from the cell body.

– **Dendrites** transmit nerve impulses towards the cell body.

This allows for the one-way flow of nerve impulses through the neurone.

Some neurones have **myelin sheaths** allowing for the faster transmission of nerve impulses.

Neurons generate and carry nerve impulses which are **electrochemical signals** that are transmitted across distances between tissues and organs. Junctions between nerve cells are called **synapses** where chemical **neurotransmitters** are secreted to transfer impulses from one neurone to the next. Nerve impulses rely on the movement of sodium and potassium ions across the membrane by passive and active transport to reset the membrane.

Nerve tissue makes up the nervous systems.

The brain and spinal cord also have **glial cells** which provide physical and chemical support to neurons and maintain their environment. The myelin sheath around the axons of some neurones is made up of Schwann cells.

Motor neurones

Motor neurons are responsible for sending impulses **from** the spinal cord and brain **to** all the muscles of the body.

Sensory neurones

Sensory neurones are responsible for generating impulses **from** the receptors that are present in the muscles, skin and other internal organs and direct them **to** the brain and spinal cord.

STEM CELLS

Stem cells are **undifferentiated** cells from which all other cells with specialised functions are generated.

To be considered a stem cell, cells must have two characteristics:

- the ability to replicate itself in an unlimited fashion to produce progeny exactly the same as the originating cell
- the ability to give rise to a specialised cell type (differentiates them from cancer cells).

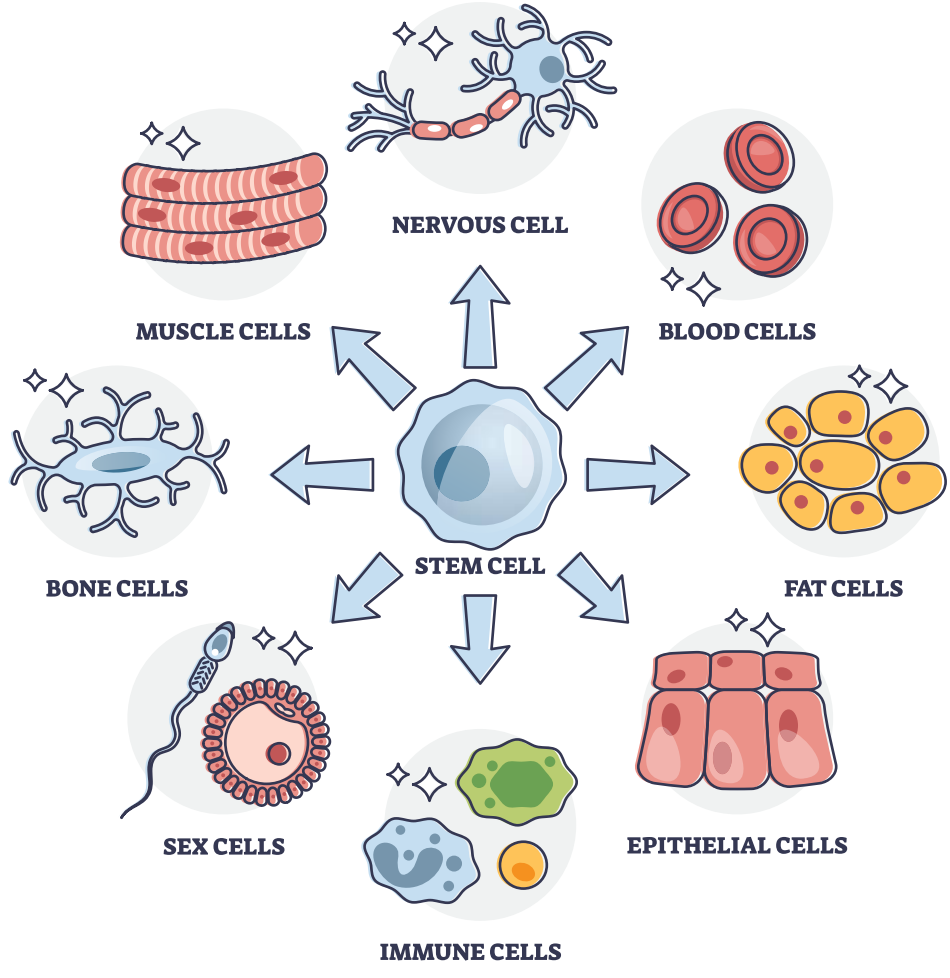


Figure 3.13: Stem cell differentiation

Levels of Potency

- **Totipotency**: the ability to produce all types of body cells. Only one example, the fertilised ovum or zygote.
- **Pluripotency**: the ability to differentiate into any of the three germ layers in the embryo: endoderm (gut, lungs, yolk sac), mesoderm (muscle, skeleton, blood vascular, urogenital, dermis), or ectoderm (nervous, sensory, epidermis), but not into extra-embryonic tissues like the placenta. Cells of the developing embryo in the blastocyst are pluripotent.
- **Multipotency**: the potential to differentiate into discrete cell types, usually of the same category e.g. epithelial cells produce other epithelial cells.
- **Unipotency**: ability to produce one type of cell e.g. surface mucous cells can only produce surface mucous cells in the lining of the stomach.

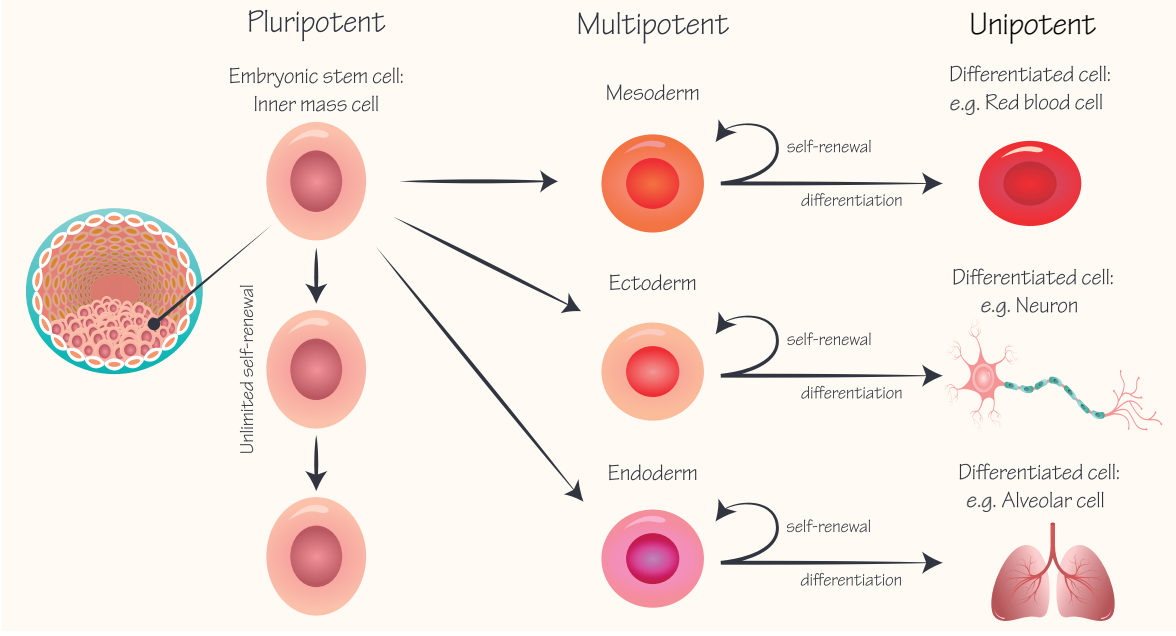


Figure 3.14: Hierarchy of stem cells

There are three types of stem cells:

- **Embryonic stem cells** come from embryos. They are pluripotent and therefore can develop into more than one type of cell in the body.
- **Adult stem cells** replace old cells and supply new cells to the growing organism. Adult stem cells are multipotent, which means they can develop into some cells of the body, but not others. For example, a stem cell from the skin (an epithelial cell) can only make skin cells or a stem cell from the blood can only make blood cells.
- **Induced pluripotent stem cells** are made in a lab. Adult stem cells are re-engineered to be stem cells using factors to change the genetics using chemicals or electrical stimulation.

Importance of stem cells in the body

- Stem cells generate replacements for cells that are lost through normal wear and tear, injury, or disease.

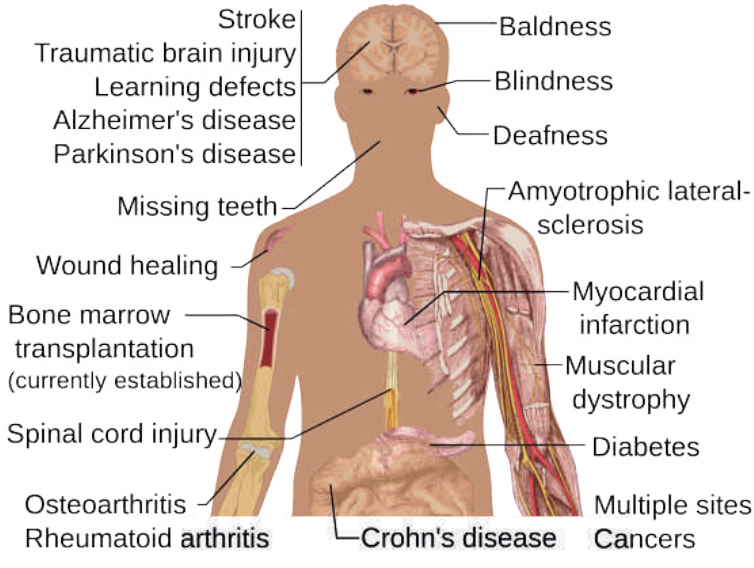
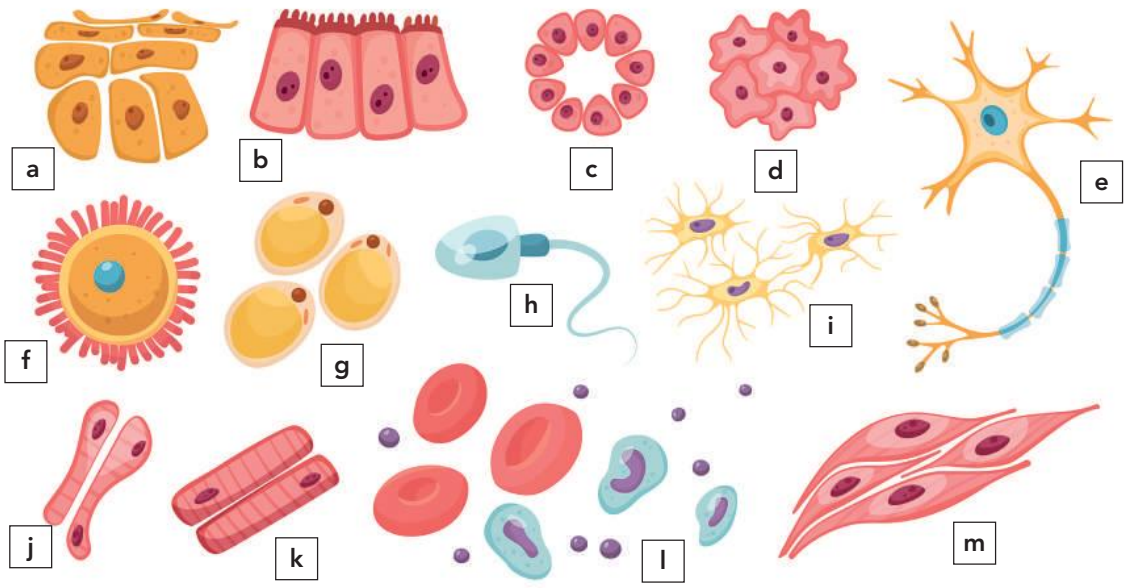


Figure 3.15: Potential uses of stem cells
From: Importance of Stem Cells | Stem Cells | University of Nebraska Medical Center (unmc.edu)

Question 1



For each of the cell types shown, state the tissue type. (1 mark each)

Cell	Tissue type	Cell	Tissue type
a		h	
b		i	
c		j	
d		k	
e		l	
f		m	
g			

Question 2

State the differences between the different types of connective tissue. (1 mark)

Question 3

Which type of tissue has a surface into space? (1 mark)

Question 4

Describe how muscle tissues are categorised into different groups. (3 marks)

Question 5

A cancer formed in the lining of the kidney tubules. Which tissue type would be affected? (1 mark)

Question 6

Nervous tissue is not completely made up of neurones. What other cells are present and describe their function. (4 marks)

Question 7

The cells forming ciliated columnar epithelial tissue have cilia on one surface. Explain their use in the location where they are found. (2 marks)

Question 8

Describe the function of smooth muscle. Explain why striated muscle would not be suitable in the locations where smooth muscle is found. (4 marks)

Question 9

Explain why blood is placed in the connective tissue group. (1 mark)

Question 10

Distinguish between a tissue and an organ. (4 marks)

Question 11

Describe the difference between pluripotent cells and multipotent cells. (2 marks)

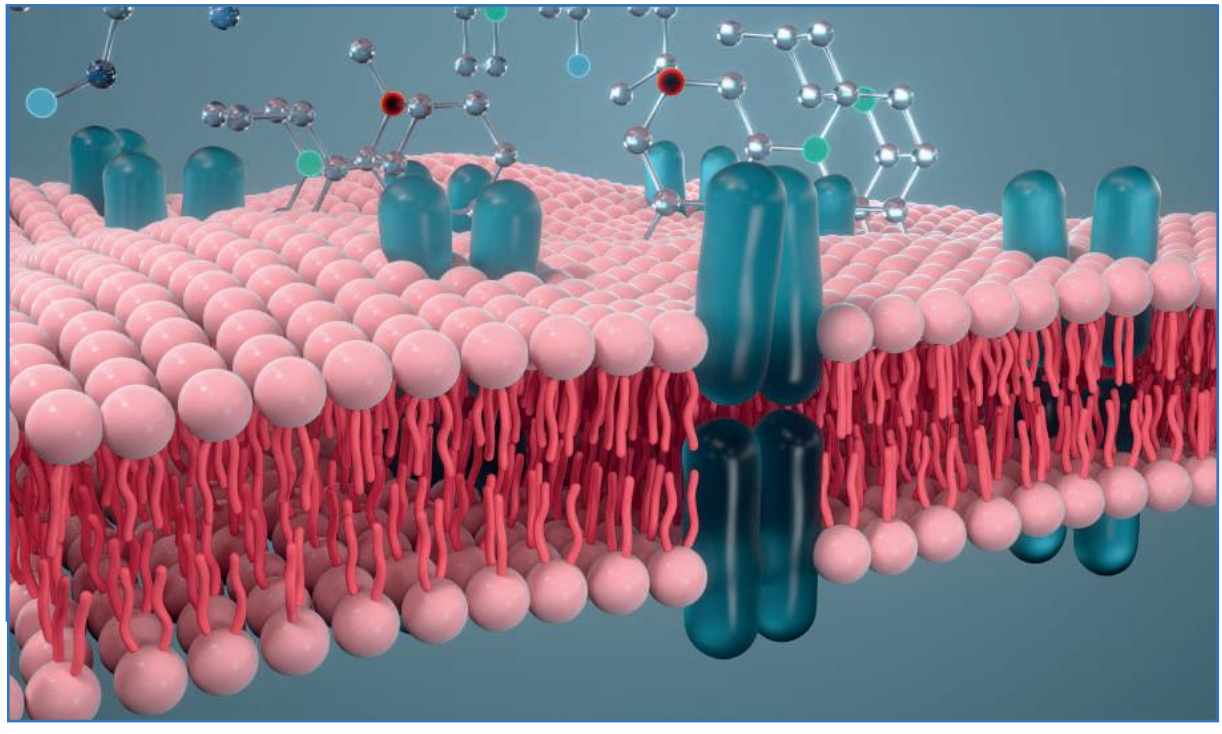
Question 12

Outline the differences between embryonic stem cells and adult stem cells. (4 marks)

Question 13

Explain the role of stem cells in in the repair of a wound such as a scraped knee. (2 marks)

4 CELL MEMBRANE AND THE EXCHANGE OF MATERIALS



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The cell membrane separates the cell from its surroundings with a structure, described by the fluid mosaic model, which allows for the movement of materials into and out of the cell by osmosis, simple diffusion, facilitated diffusion, active transport and vesicular transport (endocytosis/exocytosis).				
• Factors affecting the exchange of materials across the cell membrane include surface area to volume ratio, concentration gradients, and the physical and chemical nature of the materials being exchanged.				

CELL MEMBRANE

Without the cell membrane life as we know it would not exist. Cells can't exist without a cell membrane separating the cellular contents from the external environment.

Roles of the cell membrane

- delineates the cell from the surrounding environment
- supports the cell and helps maintain its shape; provides anchoring points for the cytoskeleton fibres
- provides protection of the contents of the cell from environmental influences
- controls the movement of materials into and out of the cell
- provides for cell recognition – can differentiate between cells of the self and foreign cells
- contains receptors for hormones or immune response chemicals

There are words to describe different fluid filled areas with respect to cells:

- intracellular – area inside the cells bounded by the cell membrane
- intercellular or interstitial – area between cells
- extracellular – fluids outside the cell which can include intercellular but also other fluids such as plasma, urine, sweat and secretions from various glands in the digestive and endocrine systems.

The current model of the structure of the cell membrane is the **Fluid Mosaic Model**, which says that the structures within the membrane can move freely within the phospholipid bilayer.

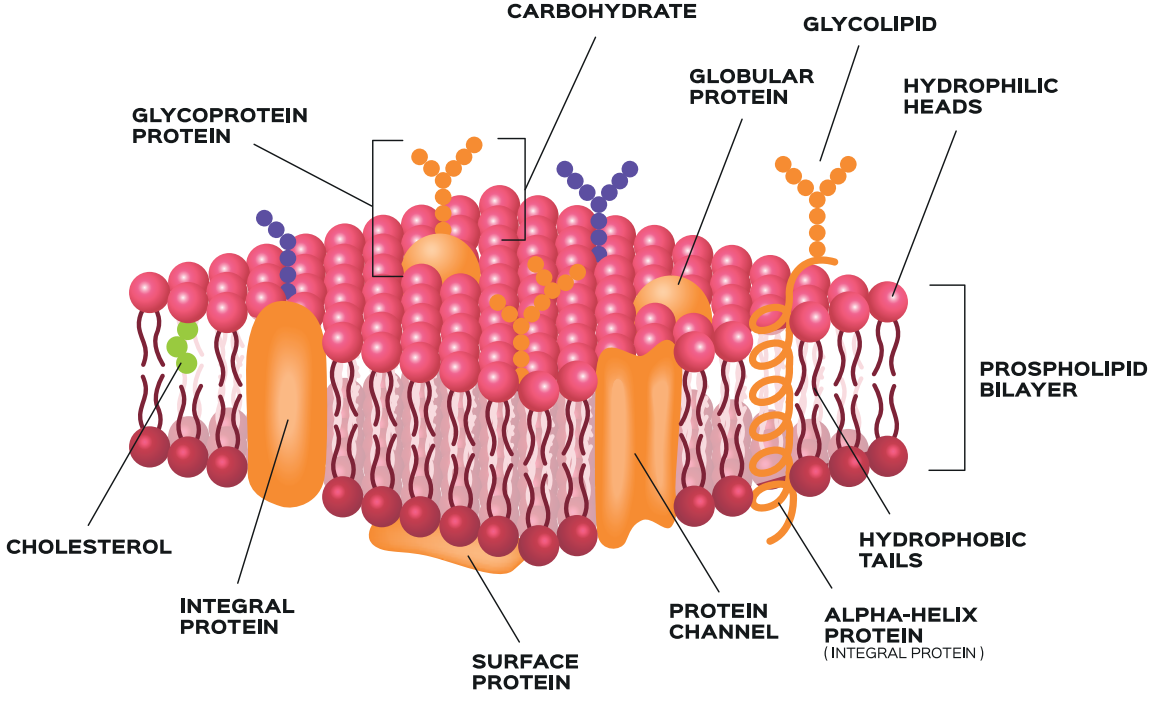


Figure 4.1: Structure of the cell membrane

Table 4.1: Components of the cell membrane, their location and functions

Component	Location	Function(s)
Phospholipids	Two layers of lipid molecules with non-polar tails adjoining	• Hydrophilic and hydrophobic sections controlling the movement of polar substances
Cholesterol	Attached between phospholipids and between the layers	• Regulate the fluidity of the membrane which is necessary to maintain its shape
Integral proteins	Embedded in the membrane across both layers	• Provide channels for transport across the membrane • Responsible for sticking cells together
Surface or peripheral proteins	On the outer surface of the phospholipid bilayer	• Involved in cell signalling – communication between cells
Glycoproteins and glycolipids	Formed by the attachment of carbohydrates to proteins or lipids within the membrane on the outside of the cell	• Structural components such as collagen • Receptors • Lubricants

Task 4.1: Label the diagram of the components of the cell membrane in Figure 4.2.

Which is the internal surface? Explain your answer.

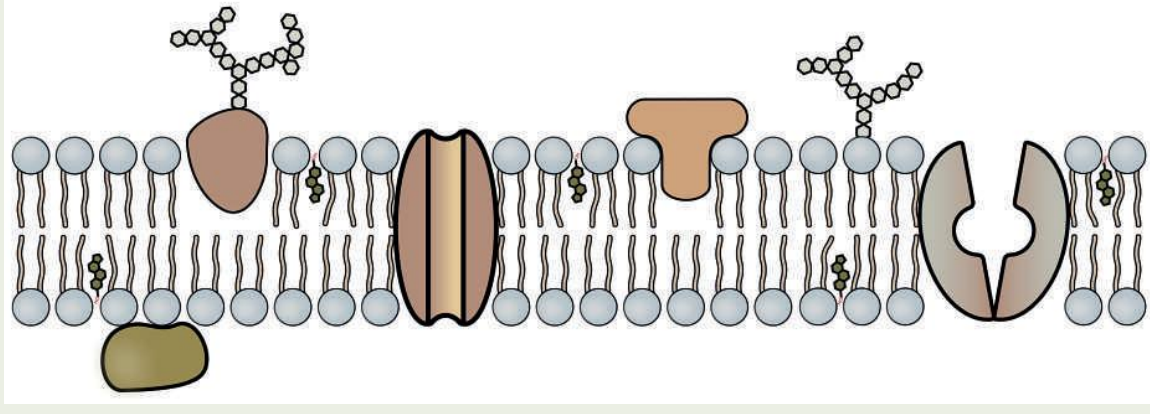


Figure 4.2: Cell membrane

EXCHANGE OF MATERIALS ACROSS THE MEMBRANE

DIFFUSION

Diffusion is the process resulting from the random motion of molecules by which there is a net movement of matter from a region of high concentration to a region of low concentration.

The movement of particles is not limited by the cell membrane.

When the concentration is equal on both sides of the cell membrane, the random movement of molecules will be equal in both directions, maintaining equal concentrations on both sides of the membrane.

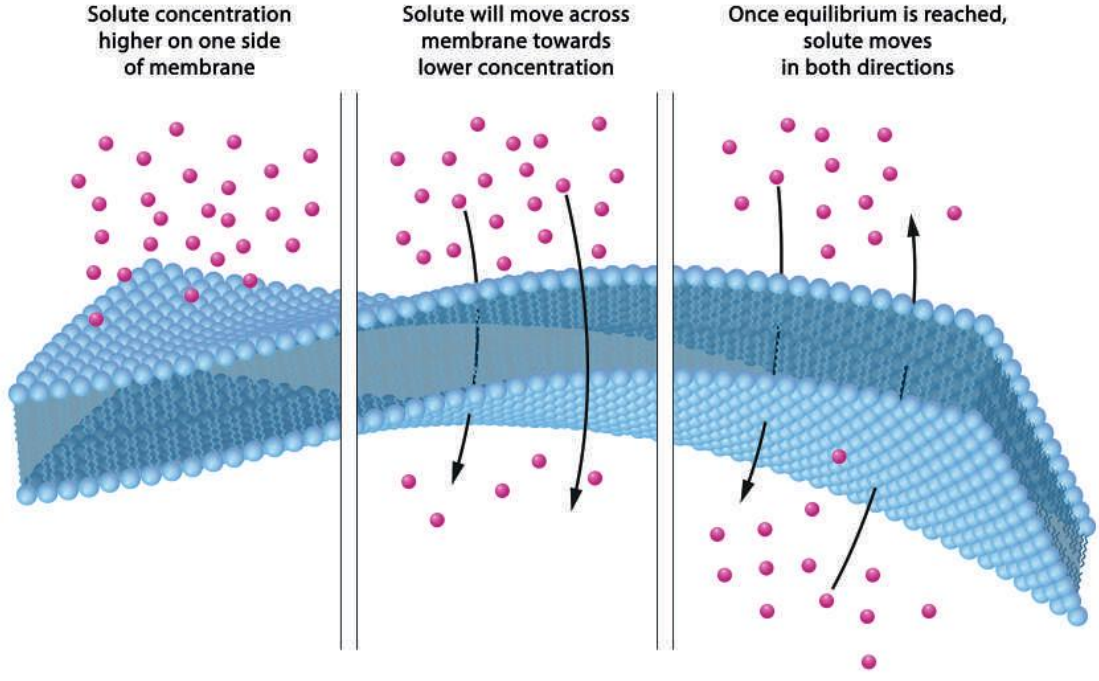


Figure 4.3: Diffusion

The movement of one type of molecule does not affect the movement of other types of molecules.

Example: the movement of carbon dioxide across the alveoli membrane of the lungs is not affected by the movement of oxygen in the other direction.

Factors affecting diffusion rates

- the greater the difference in concentration between two areas, the greater the diffusion rate
- the greater size of the molecule, the slower it will diffuse because of the difficulty of moving through the molecules of the solvent or matrix
- the greater the density of the solvent or matrix through which the molecules are diffusing, the slower the molecules will move
- the higher the temperature, the greater the rate of diffusion because molecules move faster at higher temperatures

FACILITATED DIFFUSION

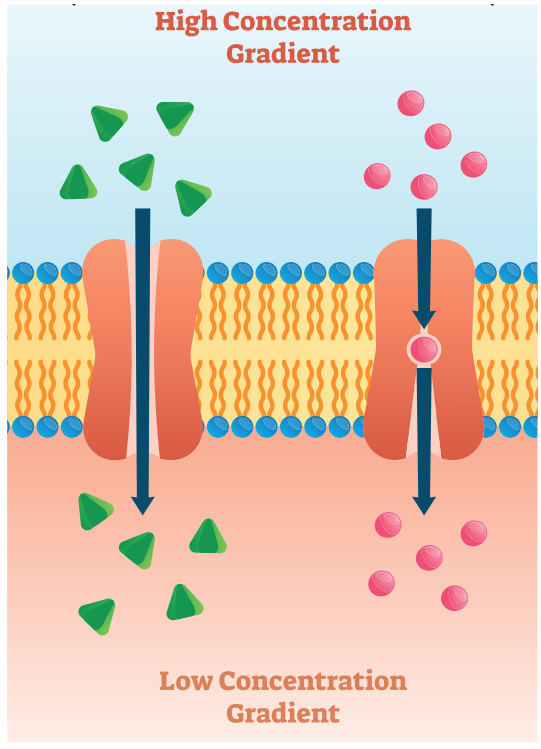


Figure 4.4: Facilitated diffusion

Facilitated diffusion is a type of diffusion in which the molecules move from the region of higher concentration to the region of lower concentration, assisted by a carrier.

- Material moves across the cell membrane with the assistance of integral proteins, sometimes called carrier proteins, down a concentration gradient (from high to low concentration) without the expenditure of cellular energy.
- The substances that move by facilitated diffusion would otherwise not diffuse easily or quickly enough across the cell membrane.
- The molecules being transported are attached to protein or glycoprotein receptors on the exterior surface of the cell membrane.
- The molecules are then passed to specific integral proteins that facilitate their passage, because channels or pores form that allow the molecules to pass through the membrane.

OSMOSIS

Osmosis is the diffusion of **water** through a semipermeable membrane according to the concentration gradient of **water** across the membrane.

Semi-permeable membrane – a membrane that allows the solvent (usually water in living cells) molecules to pass through, but not some of the solute molecules

The cell membrane is semi-permeable.

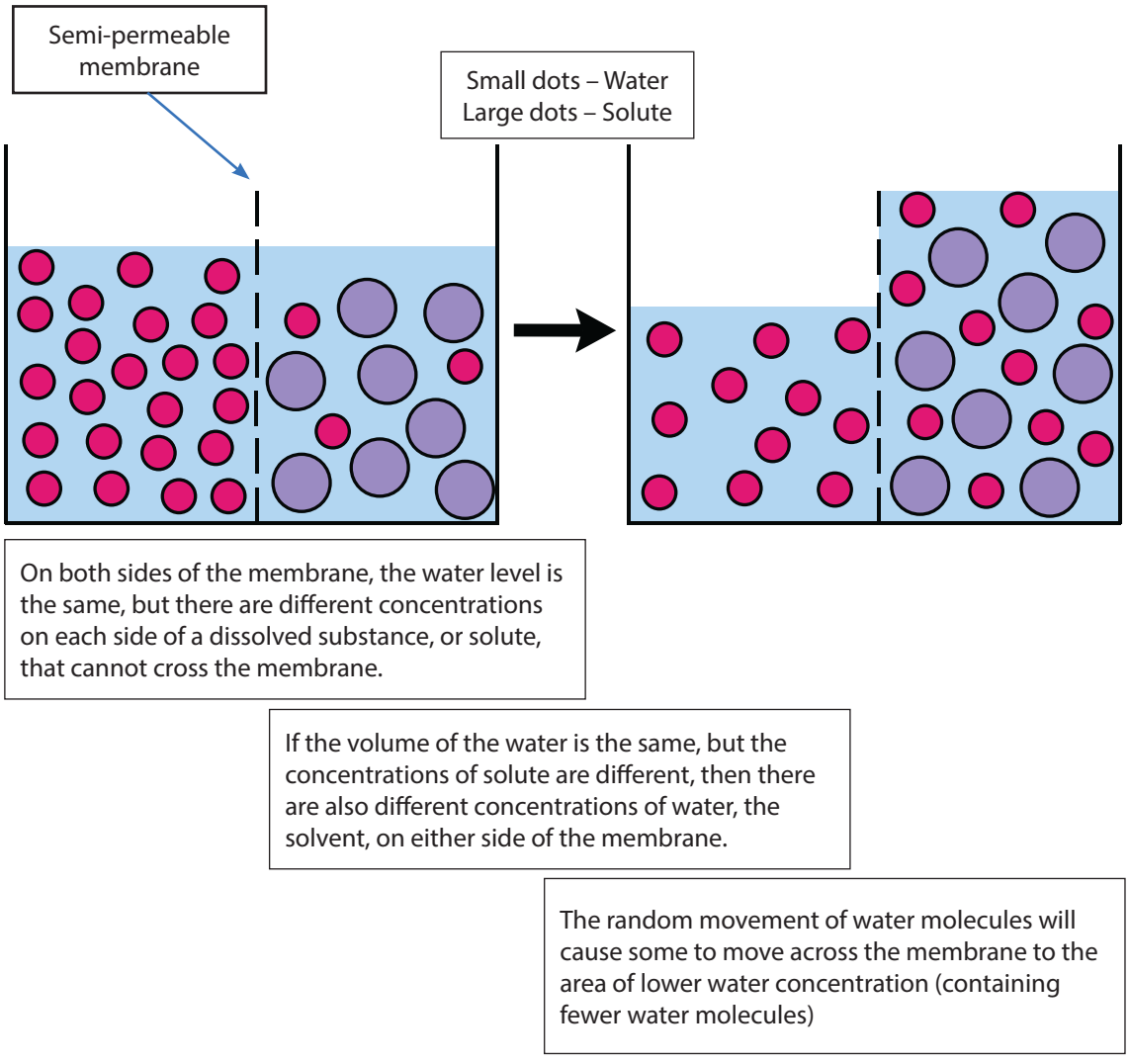


Figure 4.5: Osmosis

Factors affecting osmosis

- Pressure – by increasing the pressure on the area of high water concentration water will move more quickly across the membrane.
- Temperature – causes molecules to move faster at higher temperatures, increasing osmosis.
- Surface area – the greater the amount of surface area over which molecules can move across the membrane, the faster the rate of osmosis.
- Concentration gradient – the greater the concentration difference, the faster the rate of osmosis.

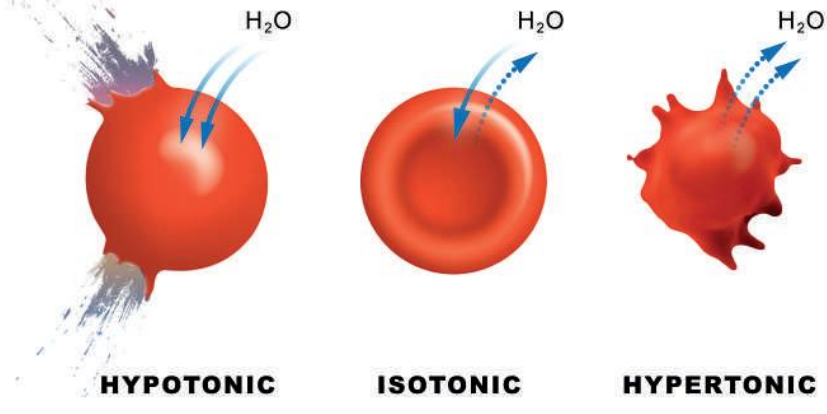


Figure 4.6: Example of the effects of osmosis

Table 4.2: Description of changes occurring in hypotonic, isotonic and hypertonic solutions

Hypotonic	Isotonic	Hypertonic
• Extracellular fluids have a lower concentration of solutes (higher concentration of water) than the intracellular fluids. • Water moves into the cells producing high enough pressure to burst the cell membrane.	• The concentration of water and solutes are the same inside and outside the cells. • Water moves into and out of the cell at the same rate and there is no change in the pressure inside the cell.	• Extracellular fluids have a higher concentration of solutes (and lower concentration of water) than inside the cells. • Water moves out of the cell, causing the cell to shrivel.

Tonicity always refers to the concentration within the cells.

ACTIVE TRANSPORT

Active transport:

- is performed by special protein channels in the cell membrane
- moves molecules against the diffusion gradient
- uses energy from the breakdown of ATP (adenosine triphosphate) to drive the movement.

Examples: uptake of glucose by epithelial cells in the villi of the small intestine; sodium-potassium ion pump in neurones.

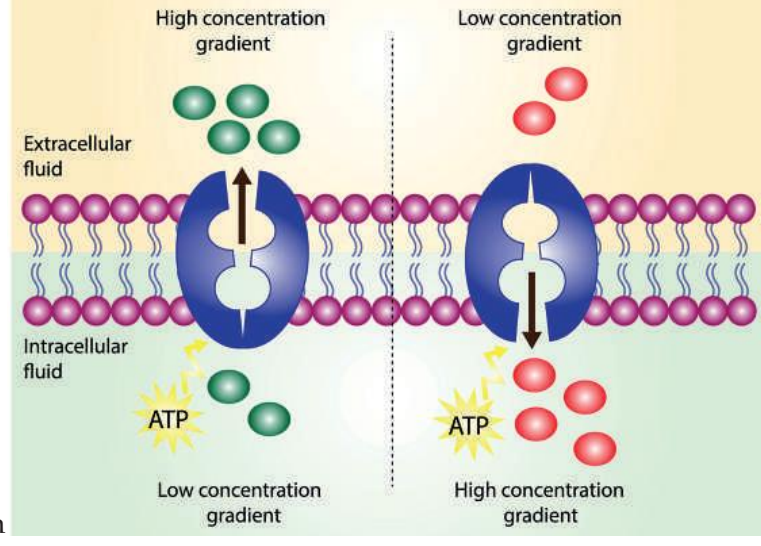


Figure 4.7: Active transport

Task 4.2: Compare diffusion, facilitated diffusion, osmosis and active transport by completing the following table.

	Diffusion	Facilitated diffusion	Osmosis	Active transport
Down a concentration gradient				
Against a concentration gradient				
Energy requirement				
Substances moved				
Notes				

ENDOCYTOSIS

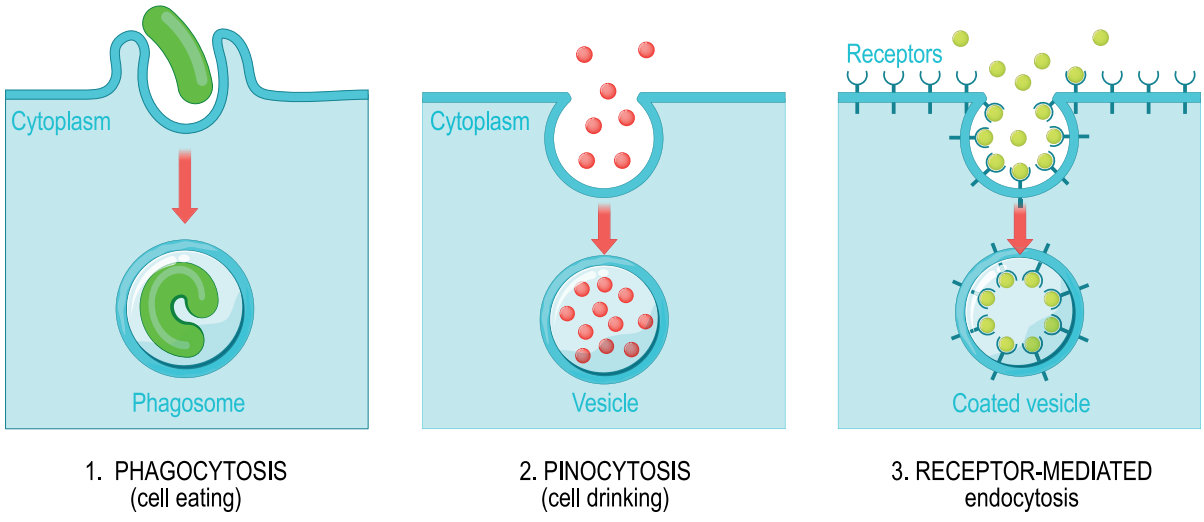


Figure 4.8: Endocytosis

Endocytosis involves the intake of materials through folding of the membrane to **encapsulate materials** that are **too large** or are **required in large amounts** and taken into the cells in the form of vesicles. These vesicles are then broken down by the enzymes in the lysosomes to release the materials into the cell.

EXOCYTOSIS

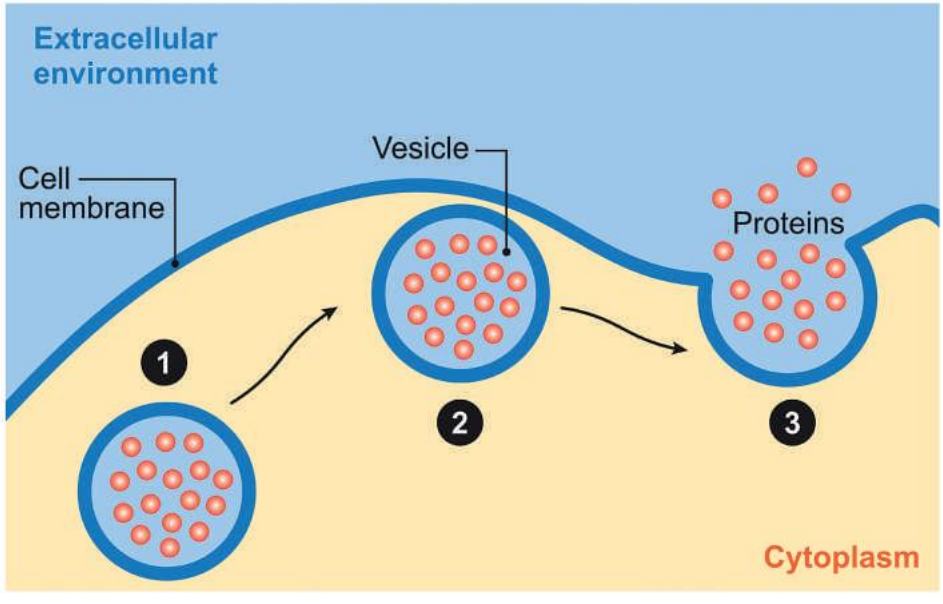


Figure 4.9: Exocytosis

Exocytosis is the opposite action to endocytosis. Vesicles containing cellular secretions formed by the Golgi bodies move towards the cell membrane where they fuse with it causing the rupture of the vesicles, releasing the contents outside the cell.

Question 1

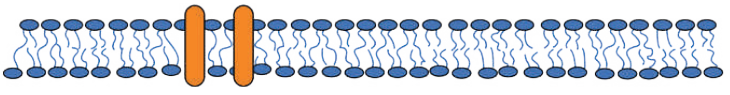
The development of the model of the cell membrane occurred over many decades. Explain why each of the following steps in development were discarded.

(a) Phase 1: shown as a solid, but flexible line surrounding the cell. (3 marks)

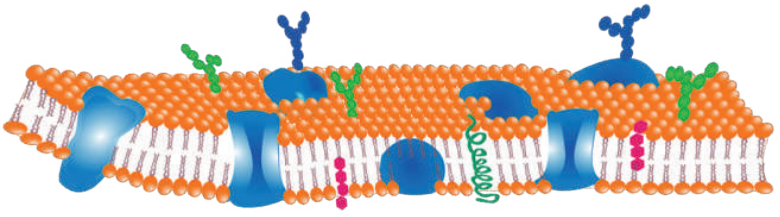
(b) Phase 2: shown as a solid, flexible line but with small gaps or pores. (3 marks)

(c) Phase 3: some of the gaps or pores in the solid, flexible line were given 'gates'. (3 marks)

(d) Phase 4: the solid line had become a double layer of phospholipids, but still have some gaps with 'gates'. (2 marks)



(e) Phase 5: the Fluid Mosaic Model where the gaps and gated gaps could move about the membrane, and proteins and carbohydrate chains are shown on either or both the internal and external sides of the membrane. (3 marks)



Question 2

Explain how the movement in diffusion and osmosis relies on the kinetic theory that a fundamental property of matter is that all molecules are in motion at all times. (3 marks)

Question 3

Use the kinetic theory to explain why diffusion would occur at a faster rate at higher temperatures. (6 marks)

Question 4

Describe the properties of a semipermeable membrane and how it helps the cell maintain its contents at optimum levels. (6 marks)

Question 5

What materials or particles are engulfed through endocytosis? (2 marks)

Question 6

By what process does the skin secrete sweat and sebum? (1 mark)

5 METABOLISM

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Biochemical processes, including anabolic and catabolic reactions in the cell, are controlled in the presence of specific enzymes.				
• Cellular respiration occurs, in different locations in the cytosol and mitochondria, to catabolise organic compounds, aerobically or anaerobically, to store energy in the form of adenosine triphosphate (ATP).				

METABOLISM

Metabolism refers to the sum total of all chemical changes that occur in the cell or body.

Metabolic rate is a measure of the amount of energy expended by a person in a given time period.

Metabolic rate is measured by:

- oxygen consumption
- carbon dioxide production
- heat production.

Basal metabolic rate (BMR) is the rate of energy use to maintain homeostasis in a resting body.

Task 5.1: Outline how oxygen consumption or carbon dioxide production can be measured to determine metabolic rate.

Energy is required by the body for:

- cell division, growth and reproduction
- active transport
- maintenance of a constant body temperature
- propagation of nerve impulses
- muscle contraction
- protein synthesis.

Factors affecting metabolic rate include:

- body size
- level of activity
- composition of the body – muscle uses more energy than fat
- diet
- growth
- gender
- genetic disposition
- hormonal or nervous conditions
- environmental temperature
- infection or illness
- use of drugs
- pregnancy.

Metabolism is made up of two processes:

- **Anabolism** requires the input of energy: it is the building up of complex molecules from simpler ones.
- **Catabolism** releases energy: it is the breaking down of complex molecules to simpler ones.

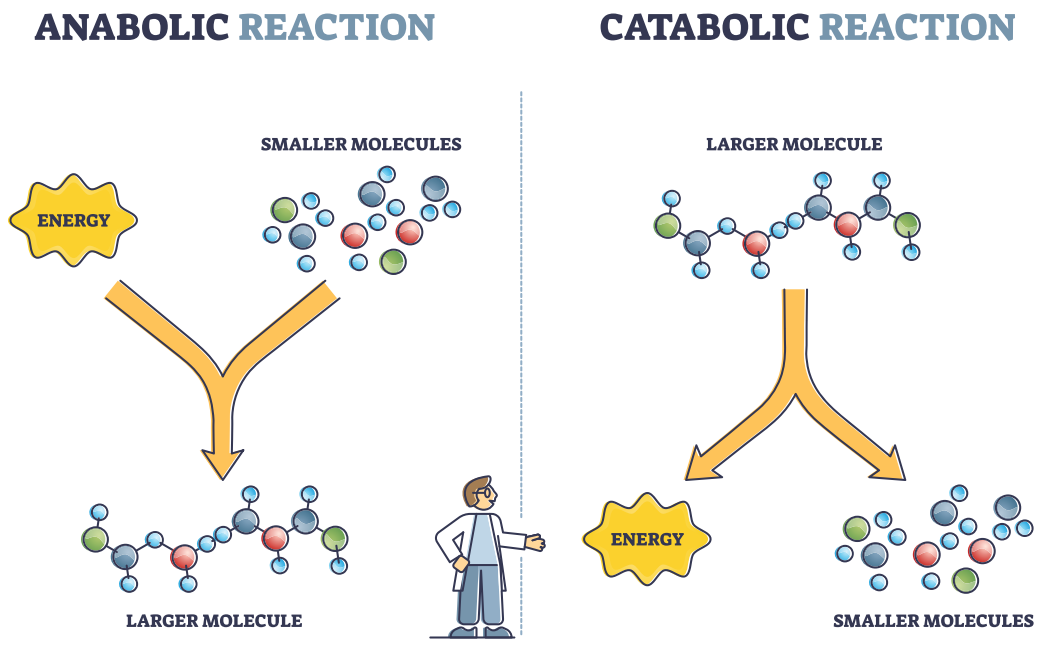


Figure 5.1: Two processes of metabolism

CELLULAR RESPIRATION

Cellular respiration refers to the chemical reaction in cells where food molecules are broken down to release energy. It is the **only** source of useable energy for cell use.

Not to be confused with:

- **gas exchange** – movement of gases across the membranes of the alveoli in the lungs
- **breathing** – movement of the chest to move air into and out of the lungs
- **artificial respiration** – ventilation of the lungs by external means such as that used in CPR.

Cellular respiration is the **only** reaction in the cell that uses oxygen and produces carbon dioxide, therefore, measuring the change in these chemicals can be a measure of the rate of respiration and therefore of metabolic rate.

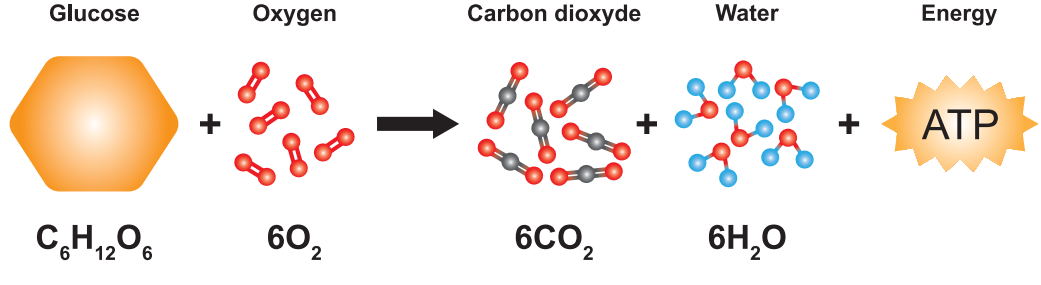


Figure 5.2: Cellular respiration

Cellular respiration takes place in many smaller steps in the body cells not as one single, simple reaction as shown in Figure 5.2.

Task 5.2: The reaction shown in Figure 5.2 indicates an overall respiration reaction. This is the same reaction for when glucose is burned e.g. when spilt onto a hot barbeque plate. Explain why cellular respiration needs to take place as series of smaller reactions rather one overall reaction.

Glucose contains chemical energy in the bonds between the atoms in the molecules.

When the bonds are broken, the energy is transferred to **chemical energy** in the ATP molecules. The energy needs to be released in amounts that can be used to produce ATP from ADP (adenosine diphosphate), but not enough to raise the temperature of the cell above optimum levels.

ATP molecules have three phosphate groups attached to a backbone of adenosine. Attaching the third phosphate group requires a lot of energy. When ATP is broken down to ADP, the last phosphate group is removed and the energy stored in the chemical bond is released for cellular use. The is the **ONLY** source of **useable** energy in cells.

(tri – three; di – two)

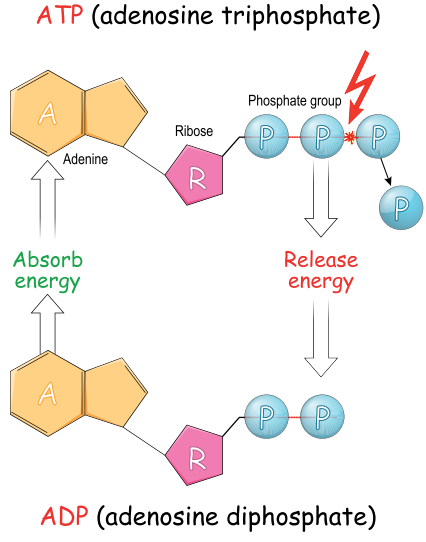


Figure 5.3: Energy changes in the production and breakdown of ATP

The reactions involved in cellular respiration occur in the cytoplasm and in the mitochondria.

- The first steps occur in the **cytoplasm**. Glucose is broken down to pyruvate in a process called glycolysis (lysis – means breakdown) which releases enough energy to produce 2ATP molecules.
- The steps breaking down pyruvate occur in the **mitochondria** where complex reactions involving electron transfer and the citric acid cycles release more energy in small packets to produce another 36 ATP molecules and release carbon dioxide and water.

AEROBIC AND ANAEROBIC RESPIRATION

Cellular respiration can occur in cells in the **presence** or **absence** of oxygen.

Aerobic – requires oxygen to undergo the complete breakdown of glucose to water and carbon dioxide.

Anaerobic – no oxygen available so only the glycolysis part of the respiration reaction occurs.

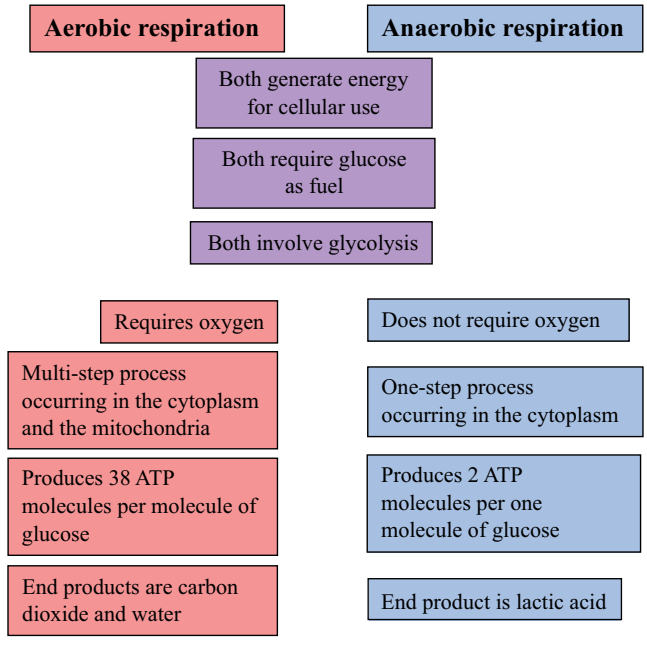


Figure 5.4: Aerobic compared to anaerobic respiration

Task 5.3: Under what body conditions would anaerobic respiration occur in cells?

USES OF ENERGY IN THE BODY

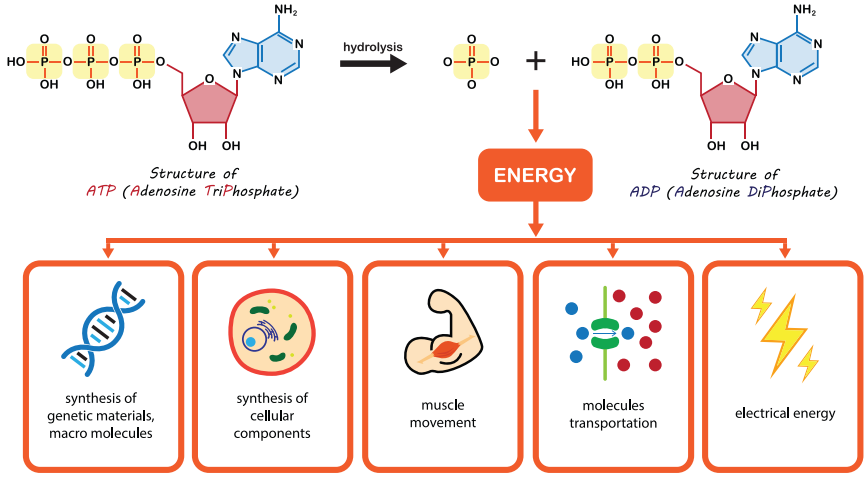


Figure 5.5: Uses of energy in the body

METABOLISM AND WEIGHT

Basal metabolic rate (BMR) is the amount of energy required, when resting, for basic life-sustaining functions like breathing, circulation, nutrient processing, and cell production.

Energy is measured in kilojoules (metric units and used in Australia) or kilocalories (imperial units and used in USA).

One kilocalorie is the same amount of energy as 4.2 kilojoules.

A kilocalorie (kcal) is defined as the amount of energy required to raise the temperature of 1 kg of water (1 litre), by 1 degree Celsius. By calculation, 1 kilojoule would raise 1 kg (1 litre) of water by 0.24 degrees.

Two other things, besides BMR, decide how many kilojoules a body uses each day:

- **How the body uses food**. Digesting, absorbing, moving and storing food all use energy. About 10% of energy intake is used for digesting food and taking in nutrients. This can't be changed much.
- **How much a body moves**. Any movement, such as playing soccer, walking to school or chasing the dog, makes up the rest of the kilojoules of energy intake in the day. This can be changed a lot, both by doing more sustained exercise or just moving more during the day.

To maintain your weight:

ENERGY IN = ENERGY OUT

No energy from food intake needs to be stored as fat, and no energy from stored fats is used.

Energy imbalances:

- Weight gain

ENERGY INPUT is **GREATER** than ENERGY OUTPUT

- Weight loss

ENERGY INPUT is **LESS** than ENERGY OUTPUT

Over 50% of Australian adults are either overweight or obese

About 25% of Australian children are overweight or obese.

Overweight and obesity are defined as the degree of abnormal or excessive fat accumulation that may impair health.

BODY MASS INDEX (BMI)

BMI provides the most useful population-level measure of overweight and obesity as it is the same for both sexes and for all ages of adults. However, it should be considered a rough guide because it may not correspond to the same level of body fat in different individuals due to build and state of health.

BMI is a simple index of weight-for-height that is commonly used to classify overweight and obesity in adults. It is defined as a person's weight in kilograms divided by the square of the person's height in metres (BMI = kg/m^2).

For adults, WHO (World Health Organisation) defines overweight and obesity as follows:

- overweight is a BMI greater than or equal to 25; and
- obesity is a BMI greater than or equal to 30.

Task 5.4: Calculate your BMI and compare it to the WHO standards.

Figures 5.6 and 5.7 indicate the problems and diseases caused by obesity.

Figure 5.6: Metabolic diseases caused by obesity

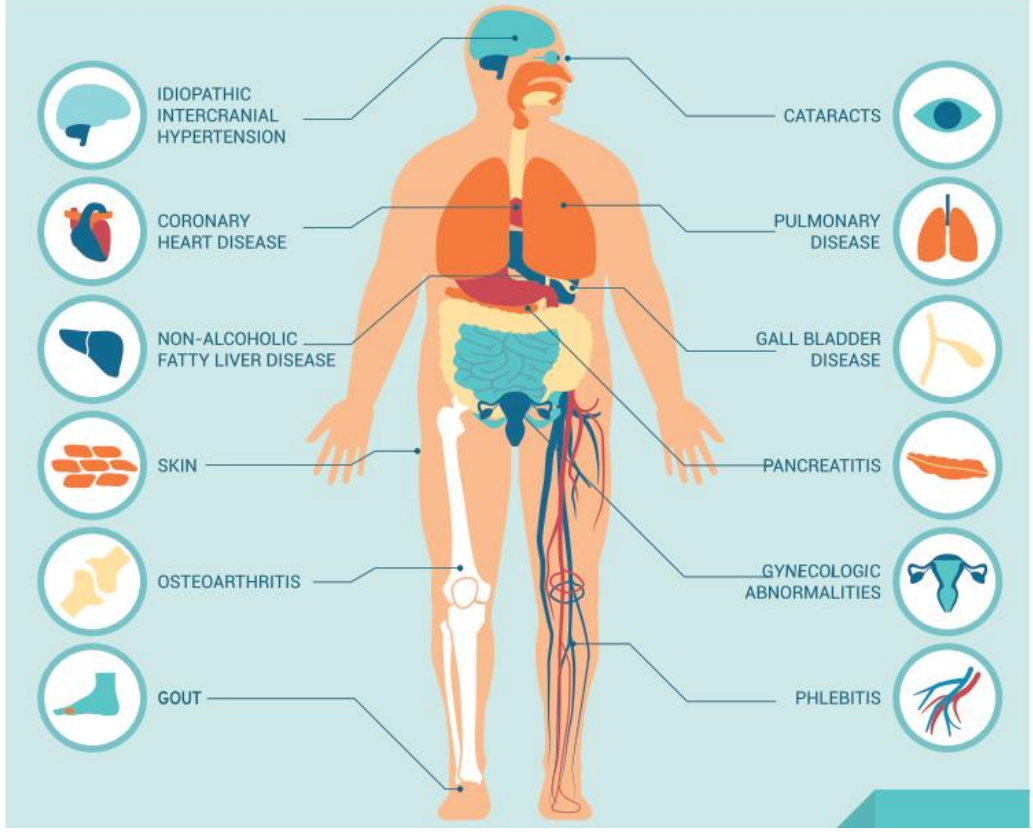


Figure 5.7: Organs and problems caused by obesity.

Figure 5.8: Three simple rules to fight obesity

Diet controls the energy intake; exercise controls the energy used by the body and together these control body weight.

Task 5.5: Comment on the impact of the lifestyle choice of being active or sedentary on the risks of developing obesity.

Question 1

The structure of mitochondria help to maximise the rate of aerobic respiration. Use Figure 5.9 to help explain why the internal structure of mitochondria is important. (3 marks)

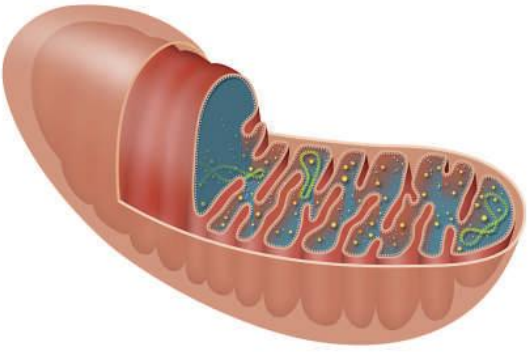


Figure 5.9: Internal structure of a mitochondrion

Question 2

Explain the differences between metabolic rates of:

(a) growing adolescents and adults (2 marks)

(b) current netball players and retired netball players (2 marks)

(c) pregnant and non-pregnant women. (2 marks)

Question 3

Describe how is energy stored in the body. (2 marks)

Question 4

The liver uses a significant amount of energy to maintain body temperature. What does this indicate about the rate of metabolic activity in the liver? (2 marks)

Question 5

Blood supply to the brain does not change during exercise, but the amount supplied to active muscles during strenuous exercise is increased by almost 10 times. Explain why this difference exists. (5 marks)

Question 6

Explain why the metabolic rate increases when the body is exposed to very high and very low temperatures. (4 marks)

6 FOOD TYPES AND DIETS

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• For efficient metabolism, cells require oxygen and nutrients, including carbohydrates, proteins, lipids, vitamins and minerals.				
• Lifestyle choices, including being active or sedentary, the use of drugs and type of diet, can compromise body functioning in the short term and may have long-term consequences.				

Food is the **source of energy** for metabolic activity in all animals, including humans.

Food can be classified according to its **chemical content and form**.

MACRONUTRIENTS

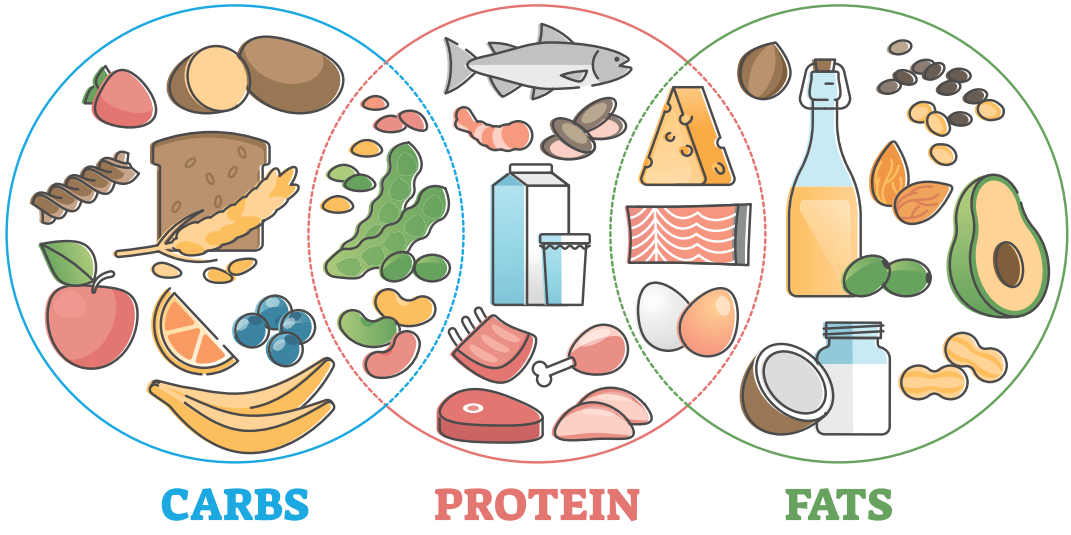


Figure 6.1: Macronutrient groups

Table 6.1: Elements contained in macronutrients

Macronutrient	Elements contained
Proteins	Carbon, hydrogen, oxygen, nitrogen, sometimes sulfur and phosphorus
Carbohydrates	Carbon, hydrogen and oxygen (in the ratio of CH_2O)
Lipids	Carbon, hydrogen and oxygen (with far fewer O than C or H)

Table 6.2: Units and sources of macronutrients

Macronutrient	Basic units	Sources
Proteins	Amino acids	
Carbohydrates	Monosaccharides	
Lipids (often referred to as fats)	Glycerol and fatty acids	

Task 6.1: Use the information in Figure 6.1 to complete Table 6.2.

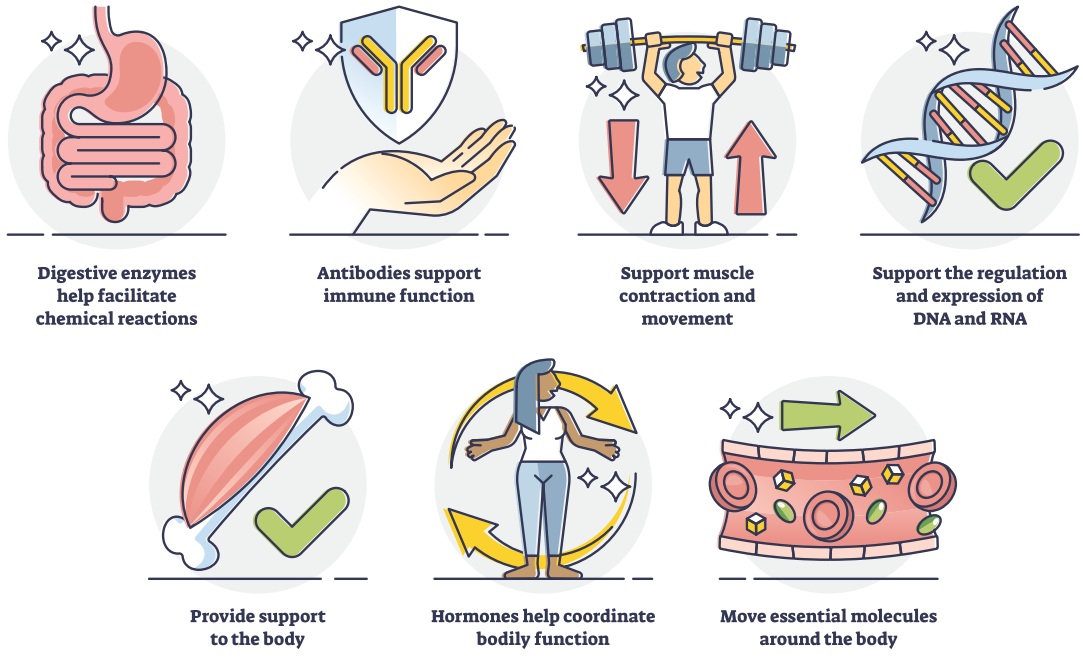


Figure 6.2: Functions of proteins

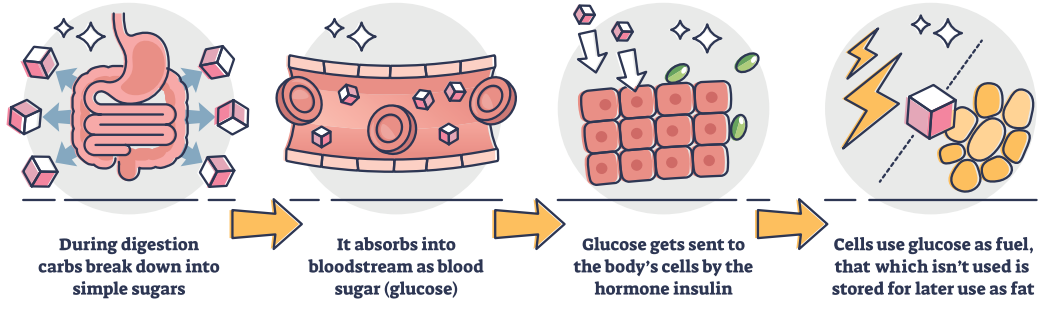


Figure 6.3: Functions of carbohydrates

Task 6.2: List the uses of lipids in the body.

Table 6.3: Testing contents of food

Food materials	Test	Colour of reagent	Positive result
Protein	Biuret test	Blue	Purple
Carbohydrate Sugars Starch	 Benedict's test Iodine test	 Light blue Orange-brown	 Green to orange-red Blue-black colour
Lipid	Brown paper	Opaque	Transparent patch
	Ethanol	Colourless	Cloudy

MICRONUTRIENTS

Figure 6.4: Micronutrient groups

Task 6.3: Use information from Figure 6.4 to complete Table 6.4.

Table 6.4: Micronutrients and sources in the diet and deficiency diseases of each

Micronutrient	Sources in the diet	Deficiency disease or symptom
Vitamins		
A		night blindness, dry skin and hair
B		fatigue, neurological disorders, heart and circulation issues
C		scurvy
D		weak, brittle bones; muscle weakness
Mineral		
Iron (Fe)		anaemia
Calcium (Ca)		rickets
Potassium (K)		heart arrhythmia
Magnesium (Mg)		muscle spasms
Iodine (I)		goitre
Zinc (Zn)		impaired growth
Selenium (Se)		thyroid dysfunction
Sodium (Na)		kidney failure

WATER AND FIBRE

Uses of water in the body include:

- regulating body temperature
- moistening tissues in the eyes, nose and mouth
- helping to maintain the flexibility of body organs and tissues to cope with internal movements
- helping blood flow to carry nutrients and oxygen to cells
- helping to lubricate joints
- increasing the efficiency of the kidneys and liver by flushing out waste products
- helping the movement of contents in the large intestine and defaecation
- dissolving minerals and nutrients to make them accessible to your body.

Sources of water include:

– drinking of fluids
– water in foods
– metabolic water formed from the respiration reaction in cells.

Dietary fibre is of two different types.

- **Soluble fibre** helps to slow the emptying process in the stomachs, which helps you feel fuller. It also helps to lower cholesterol and stabilise blood glucose levels. It is found in plant foods such as fruits, vegetables, oats, barley and legumes.
- **Insoluble fibre** absorbs water to help soften the contents of the bowels and support regular bowel movements. It also helps us feel fuller and keeps the bowel environment healthy. It is found in wholegrain breads and cereals, nuts, seeds, wheat bran and the skins of fruit and vegetables.

DIETS

Humans evolved eating an omnivorous diet, but today many chose different diets. Within each group shown in Table 6.5, there are many variations based on the choice of ingredients.

Table 6.5: Different diets

Omnivorous	Eats both animal products and plant products
Vegetarian	Does not eat meat, but will eat animal products such as eggs and milk products, as well as plant products
Vegan	Do not eat animal products; diet is of plant products only

Task 6.4: Outline the differences for the intake of iron and proteins between omnivorous and vegan diets.

The Australian Dietary Guidelines 2013 provide specific recommendations on the amount of food to consume from each food group each day, for a balanced diet.

There are recommendations for the following groups:

- Toddlers (1–3 years)
- Children (4–11)
- Adolescents (12–18)
- Adults (19–70)
- Pregnant and breastfeeding women
- Older adults (70+).

Consuming the recommend amounts will provide you with adequate energy (kilojoules) and nutrients (vitamins, minerals etc) for your age and gender.

Question 1

Describe the difference between macronutrients and micronutrients. (2 marks)

Question 2

Explain which of the children is malnourished. (4 marks)

Question 3

Food tests were conducted on a piece bread and butter and jam. State what tests could be used and the likely results of the tests. (15 marks)

Question 4

Explain why tests for protein usually test for the presence of nitrogen. (2 marks)

Question 5

Explain why protein deficiency leads to slow growth. (2 marks)

Question 6

Explain why peeling vegetables before eating is detrimental to a balanced diet. (2 marks)

Question 7

English sailors in the 18th and 19th centuries used to be called 'limeys' because they would be given a ration of limes or other fruit or vegetables, such as onions, as part of their diet. The normal diet of other sailors was hardtack (hard cereal-based biscuits) and salted meat. Explain the likely difference in health of the sailors who had a ration of fruit and those who didn't during a long sea voyage. (4 marks)

Question 8

Explain why eating a whole fresh orange or apple is better for you than just drinking the juice from these fruits. (6 marks)

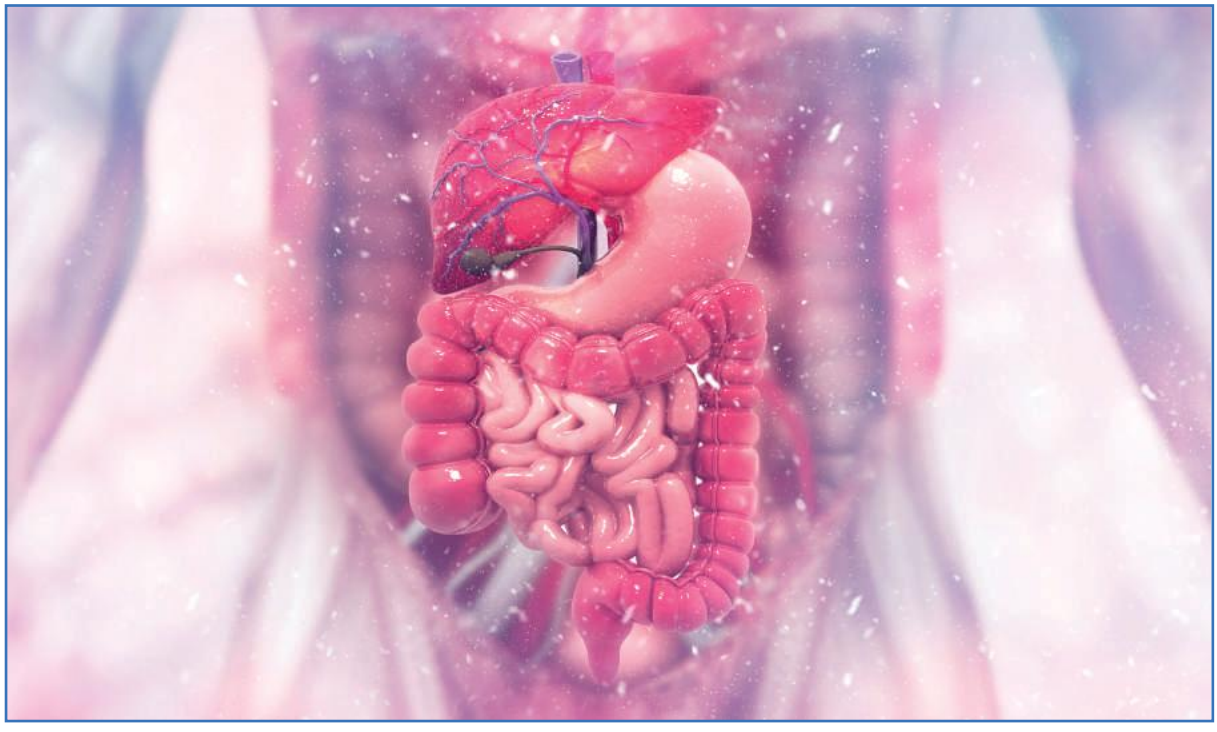
Question 9

Diets are sometimes referred to by using their place or time of origin. For example, the Mediterranean diet and the Paleo diet. Describe the general differences between these diets. (2 marks)

Question 10

Explain why there are different dietary recommendations for people of different ages or life stages. (2 marks)

7 DIGESTIVE SYSTEM



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The supply of nutrients in a form that can be used in cells is facilitated by the structure and function of the digestive system at the cell, tissue and organ levels.				
• Digestion involves the breakdown of large molecules to smaller ones by mechanical digestion (teeth, peristalsis, churning and bile) and chemical digestion (by enzymes with distinctive operating conditions and functions that are located in different sections of the digestive system).				
• The salivary glands, pancreas, liver and gall bladder produce or store secretions which aid the processes of digestion.				
• Absorption requires nutrients to be in a form that can cross cell membranes into the blood or lymph and occurs at different locations, including the small intestine and large intestine.				
• Elimination removes undigested materials and some metabolic wastes from the body.				

DIGESTION

Digestion is the process of making food available to cells by mechanically and enzymatically breaking it down into simpler chemical compounds in the digestive tract so they can be absorbed into the blood for transport and for use in cells.

Types of digestion:

- **Mechanical** – where food is broken down into smaller pieces of the same material or dissolved. There is no change in the chemical composition of the food.
- **Chemical** – where food is broken down into smaller chemical units usually with the aid of enzymes.

DIGESTIVE SYSTEM

The digestive system is made up of the alimentary canal or digestive tract and the organs that produce materials that help in the digestion of food.

Any material inside the digestive tract has not been absorbed so is considered to be outside the body, like the contents of a pipe are not part of the pipe.

The **digestive system is in sections** because each section has specific conditions and secretions that are optimal for different enzymes.

The **sequence of organs in the digestive tract** allows the sequential breakdown of food from most complex to the basic units of food.

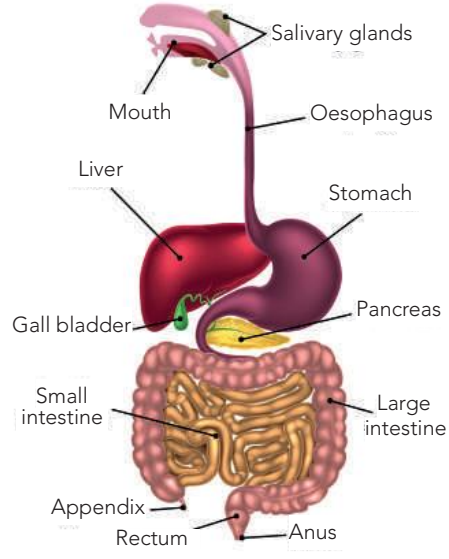


Figure 7.1: The digestive system

MOUTH

The mouth contains:

- Teeth – cut and grind food into smaller pieces for swallowing
- Tongue – moves food around the mouth for even chewing and swallowing
- Salivary glands – located in cheeks and under the tongue – produce saliva which contains enzymes (amylase) fluids to dissolve food and make swallowing easier.

Teeth

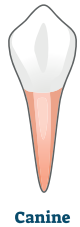
 **Incisor**	 **Canine**	 **Premolar**	 **Molar**
Incisors – shear food during biting	Canines – tearing food	Premolars – tearing and coarse grinding	Molars – crushing and grinding

Figure 7.2: Types of teeth in the human mouth

Humans have two sets of teeth. The first set is complete by the age of two (20 teeth) and are replaced between the ages six and twelve with the permanent set (32 teeth).

Tongue and taste

The tongue contains tastebuds that can distinguish five different basic flavours:

- sweet
- sour
- salty
- bitter
- umami.

Foods taste different as a result of the combinations of these five flavours as interpreted by the brain.

Myth: different areas of the tongue are sensitive to different flavours. All taste buds can detect all flavours. For further information go to https://www.bbc.com/future/article/20171012-do-our-tongues-have-different-taste-zones

SWALLOWING

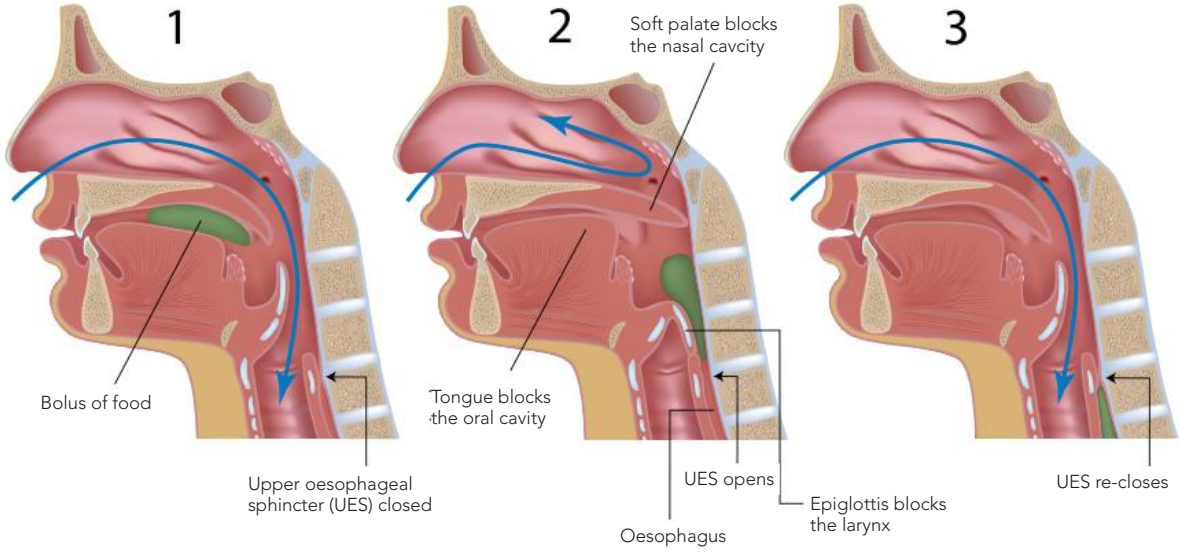


Figure 7.3: Swallowing

Swallowing is a reflex when the tongue moves food to the back of the mouth, causing the trachea and nasal cavity to be closed and the oesophagus to be opened to allow the passage of food to the stomach. The epiglottis blocks the trachea and the soft palate closes the nasal cavity during swallowing.

PERISTALSIS

Peristalsis refers to the rhythmic contraction and relaxation of the muscles of the oesophagus so that food is forced down the oesophagus to the stomach. This movement is involuntary and is necessary for the movement of food through the digestive tract. Peristalsis occurs throughout the digestive tract to keep the food moving.

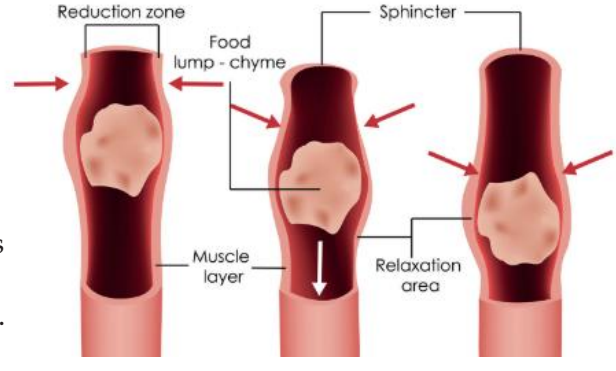


Figure 7.4: Peristalsis

STOMACH

The stomach is a muscular sac that stores food temporarily while enzymes work on the contents.

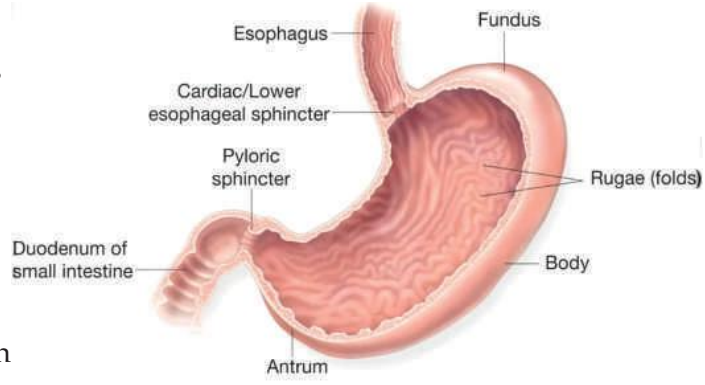


Figure 7.5: The stomach
From: https://sonographictendencies.com/2017/08/20/gastrointestinal-sonography/

Structures

- It has three layers of muscle tissue that contract to churn the contents to mix with digestive juices.
- The cardiac sphincter at the junction between the oesophagus and the stomach allows food to enter, but not return to the oesophagus.
- The pyloric sphincter controls the release of food (now called chyme) into the small intestine.
- Rugae are folds in the stomach wall that allow it to expand when food enters.
- The thick mucus-membrane lining of the walls is densely packed with small gastric glands.

Functions

- It temporarily stores food.
- Its muscles contract and relax through peristalsis to mix and mechanically break down food.
- It produces enzymes and other specialised secretions to provide the optimal conditions for the digestion of food.

Gastric glands

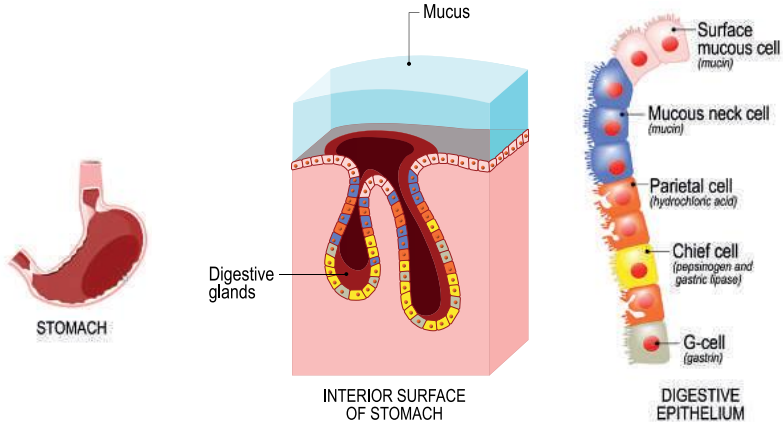


Figure 7.6: Gastric gland and cells of the epithelial lining

Gastric glands are found in the lining of the stomach and produce:

- **Gastrin** promotes the release of HCl.
- **Hydrochloric acid (HCl)** provides an acid environment for activating the pepsin enzyme.
- **Pepsinogen** is the inactive form of the enzyme pepsin.
- **Mucin** is the basis for the mucus secretion covering the cells of the stomach lining, preventing them from being digested by the active enzymes in the stomach.

See also Figure 3.4 in Chapter 3

SMALL INTESTINE

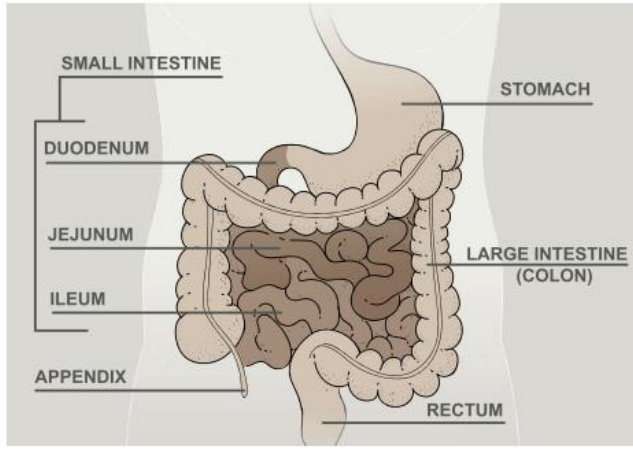


Figure 7.7: Positions of the small and large intestines

Figure 7.8 illustrates ever increasing magnifications of the wall of the small intestine showing the folds in the intestinal wall covered by villi which in turn are covered by epithelial cells that have microvilli.

The small intestine is surrounded by three different layers of muscle tissue to continue the movement of materials through the tract by peristalsis. It is about 6.7m in length.

The villi and microvilli increase the surface-area-to-volume ratio to allow for the increased efficiency of absorption of nutrients and for the movement of materials across the surface of the lining of the small intestine.

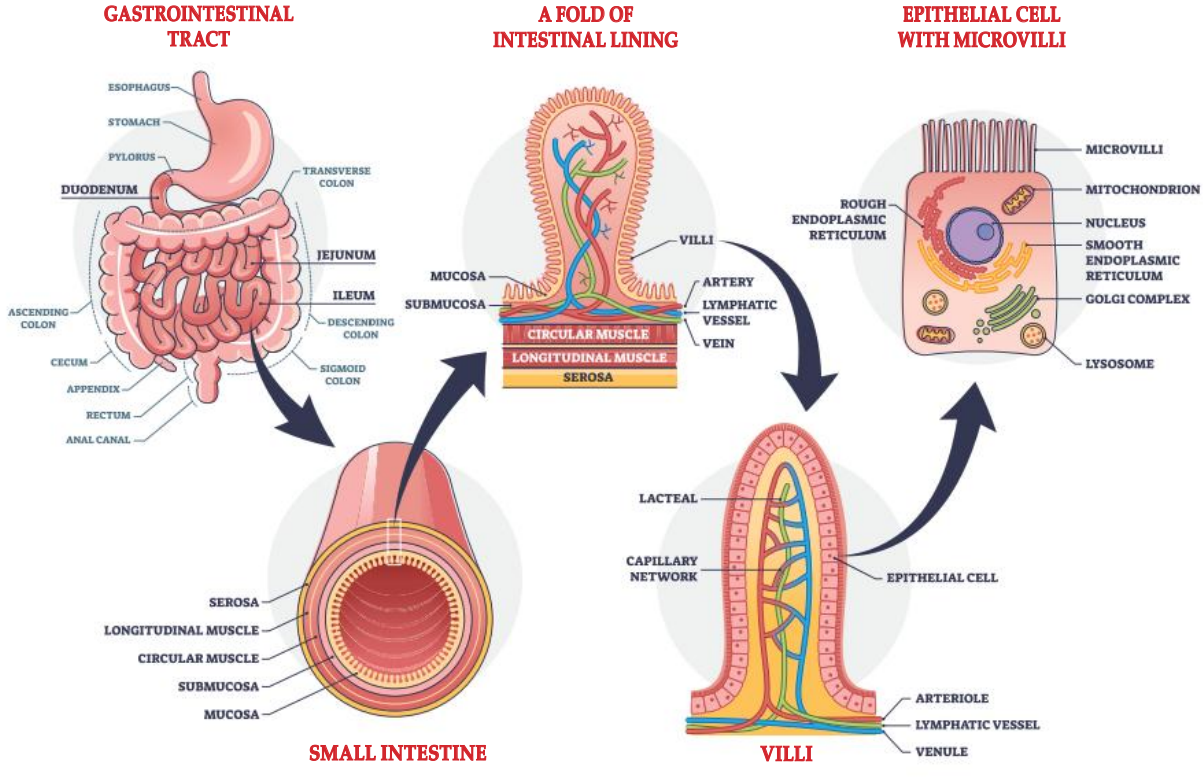


Figure 7.8: Structure of the small intestine

Materials absorbed into the blood vessels of the villi are amino acids, monosaccharides/simple sugars, vitamins, minerals and water.

Materials absorbed in to the lacteals (lymph vessels) of the villi are glycerol and fatty acids.

LARGE INTESTINE

The large intestine has a larger diameter than the small intestine but is shorter in length at about 1.5m.

The functions of the large intestine are:

- the absorption of vitamins
- the absorption of water and electrolytes
- the bacterial fermentation of indigestible materials
- the formation and elimination of faeces.

Removal of materials (faeces) from the large intestine is called elimination or defaecation is aided by peristalsis and controlled release through the anal sphincter.

Faeces contain 75% water and 25% solid matter.

The solid matter is composed of:

- bacteria (30%)
- undigested food and fibre (30%)
- fat (10%-20%)
- inorganic matter (10-20%)
- other protein (2-3%)
- carbohydrate or undigested plant matter (25%).

Faeces are brown due to the break down products of red blood cells that occurs in the liver and are stored as bile in the gall bladder.

The colour and consistency of faeces can change according to the food eaten or different health conditions.

Constipation is infrequent and/or difficult bowel movements or defaecation.

Diarrhoea is having 3 or more loose or liquid stools (poos) in one day, or defaecating more frequently than normal.

Question 1

List the organs of the digestive tract in order from entry to exit. (5 marks)

Question 2

Name the sphincters that control the movement of food through the digestive tract. (6 marks)

Question 3

Explain what happens if food enters the appendix. (4 marks)

Question 4

Sometimes just before mealtimes, your stomach 'rumbles'. Explain why this occurs. (2 marks)

Question 5

Explain why bacteria are very important in the function of the large intestine. (4 marks)

Question 6

Explain how stomach banding influences the movement of material through the digestive tract. (6 marks)

Question 7

Explain why mechanical digestion needs to occur before chemical digestion. (3 marks)

Question 8

Describe the problem caused by the contents of the stomach returning to the oesophagus. (4 marks)

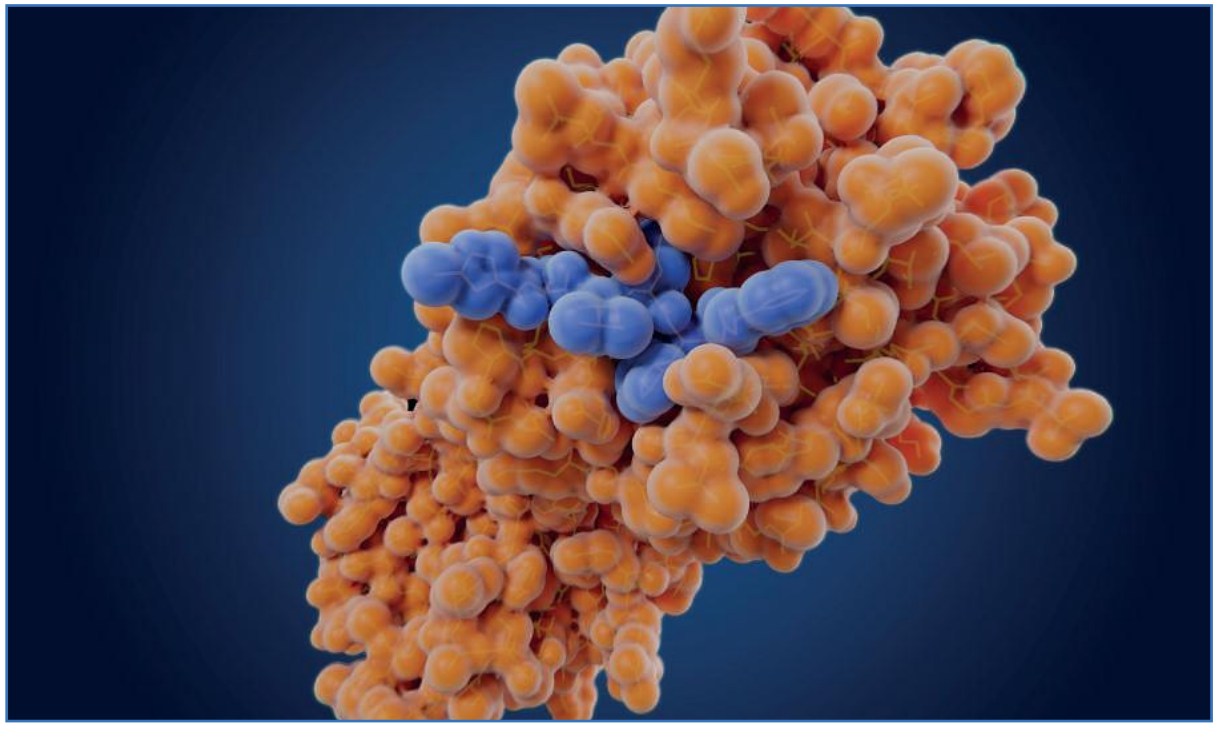
Question 9

Describe how the efficiency of absorption of nutrients is optimised by the structure of the digestive tract. (4 marks)

Question 10

As a result of bowel cancer, part of the large intestine can be removed. Describe how this would affect the functioning of the large intestine. (4 marks)

8 ENZYMES



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Biochemical processes, including anabolic and catabolic reactions in the cell, are controlled in the presence of specific enzymes.				
• Digestion involves the breakdown of large molecules to smaller ones by mechanical digestion (teeth, peristalsis, churning and bile) and chemical digestion (by enzymes with distinctive operating conditions and functions that are located in different sections of the digestive system).				
• Enzyme function can be affected by factors including pH, temperature, presence of inhibitors, co-enzymes and co-factors, and the concentration of reactants and products.				

ENZYMES

- Enzymes are proteins that help speed up chemical reactions in our bodies without being used up in the reaction. They are also known as organic catalysts.
- All chemical reactions within cells require the presence of enzymes.
- Enzymes are specific to particular reactions.
- Enzymes work on surfaces.
- Enzymes work according to the shape of the molecules in the reaction.

Catalysts/enzymes reduce the amount of energy required for a reaction to proceed (activation energy).

Reactions requiring reduced levels of activation energy can occur at lower temperatures i.e. at body temperature, without changing the amount of energy released from the reaction.

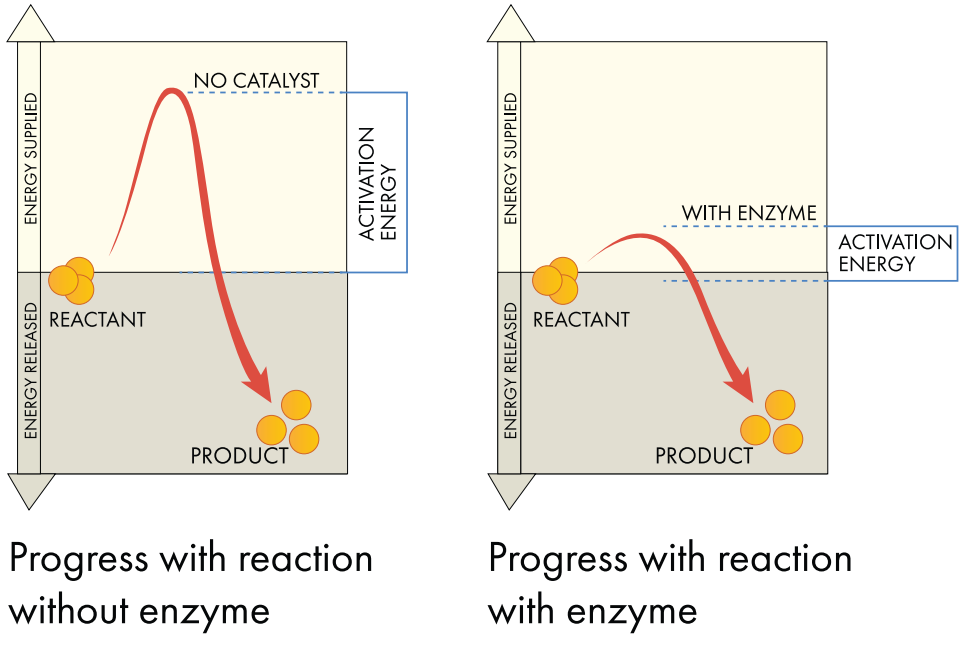


Figure 8.1: Differences in activation energy required for catalysed and uncatalysed reactions

Activation energy is the minimum amount of energy that must be provided for reactants to result in a chemical reaction.

Task 8.1: Think about the cellular respiration overall reaction and the burning of sugar. They are essentially the same reaction. One occurs at body temperature and the other requires temperatures of about 350°C to occur and releases a great amount of energy all at once. Explain how cellular respiration can occur at body temperature.

ENZYME FUNCTION

There are two models for the actions of enzymes during reactions. (Substrate is the material on which the enzyme works.)

Induced Fit Model

- The shape of the **active site changes** to fit the shape of the substrate.
- When the products are released, the active site returns to its original shape.

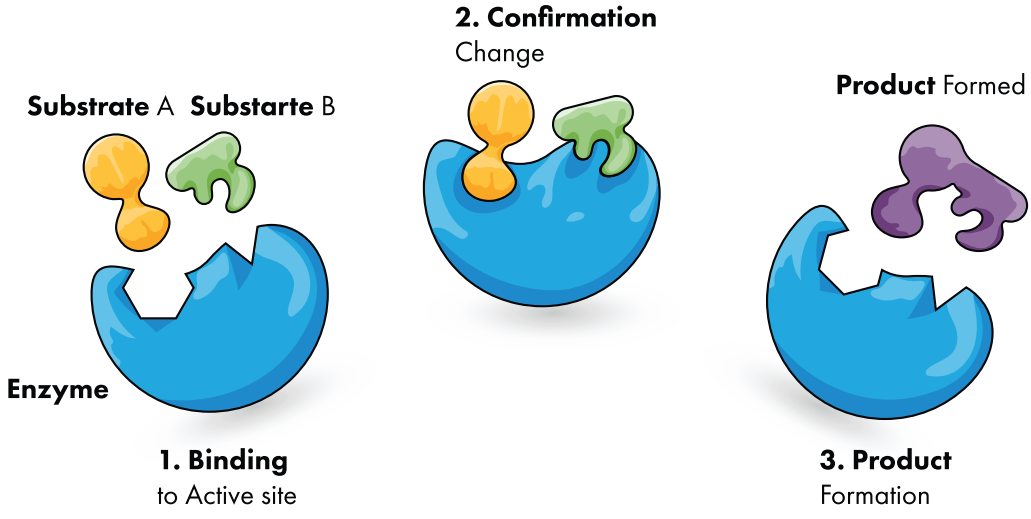


Figure 8.2: Induced fit model

Lock and Key Model

- The shape of the substrate **exactly matches** that of the active site on the enzyme.
- If the substrate does not fit exactly into the enzymes active site, no reaction will occur.

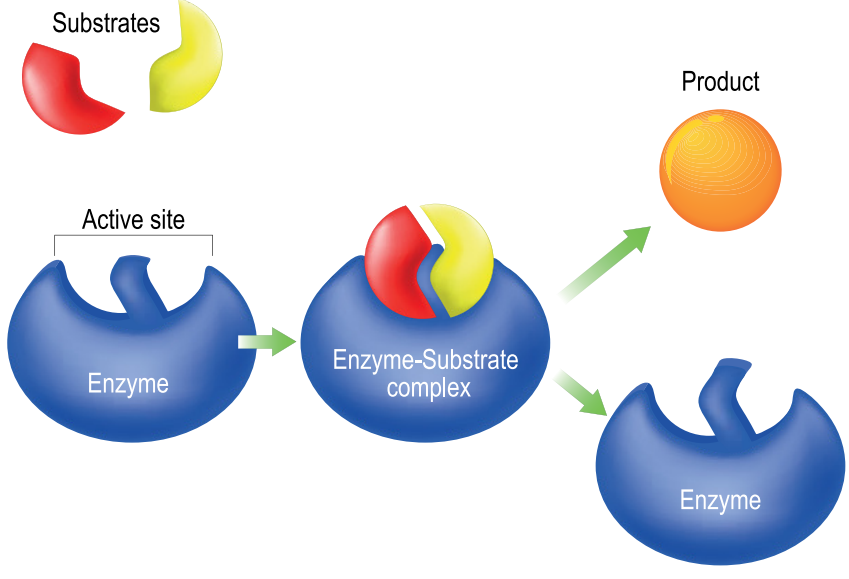
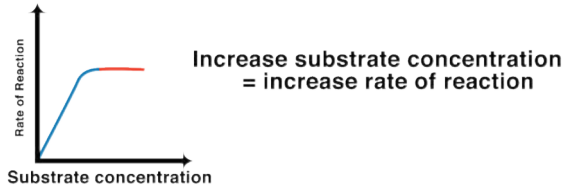


Figure 8.3: Lock and Key Model

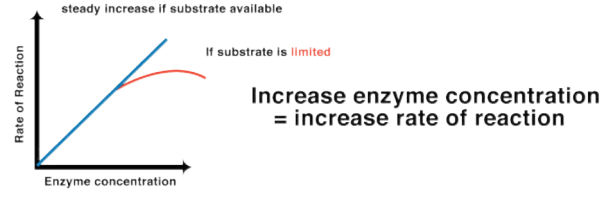
FACTORS AFFECTING THE ACTION OF ENZYMES

1. **Substrate concentration**



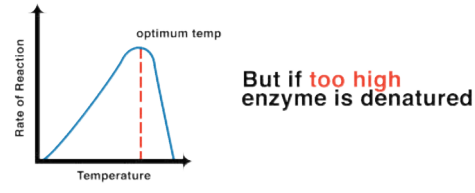
- The rate of reaction increases as the substrate concentration increases, until a limiting rate is reached when all the active sites on the enzymes are being used.

2. **Enzyme concentration**



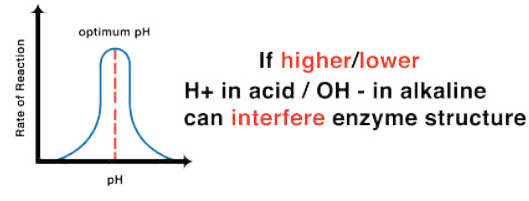
- The reaction rate increases as the concentration of the enzyme is increased, unless the amount of substrate is limited.

3. **Temperature**



- The movement of molecules increases with an increase in temperature causing an increase in the rate of reaction. After a certain temperature, however, an increase in temperature causes a decrease in the reaction rate, due to denaturation of the protein structure and disruption of the active site.
- Optimum temperature is the temperature at which the reaction rate is the greatest.

4. **pH**



- The acidity or alkalinity of the solution will change the rate of reaction.
- Each enzyme has its optimum pH at which the rate of reaction is at its greatest.

5. **Presence of inhibitors**

- Inhibitors can occupy or change the shape of the active site on the enzyme, stopping or reducing enzyme activity.

6. **Presence of co-factors and co-enzymes**

- Cofactors (e.g. iron, copper and magnesium) and coenzymes (organic cofactors e.g. B group vitamins) increase the rate of reaction by aiding energy transfer or the alignment of the substrate with the enzyme's active site.

TYPES OF ENZYMES IN DIGESTION

Enzymes are named for the substrate on which they act. Their name ends in -ase.

For example, lipase works on lipids; proteases work on proteins; amylase work on carbohydrates (amylase comes from the Latin word *amylum* meaning starch).

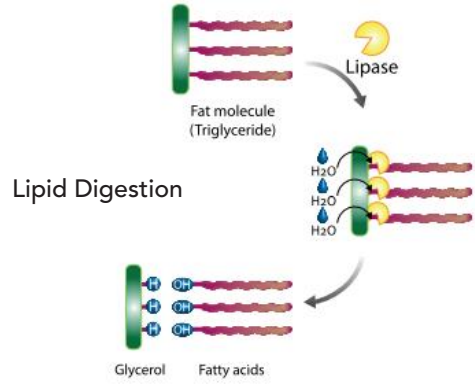


Figure 8.4: Lipid digestion – one step breakdown

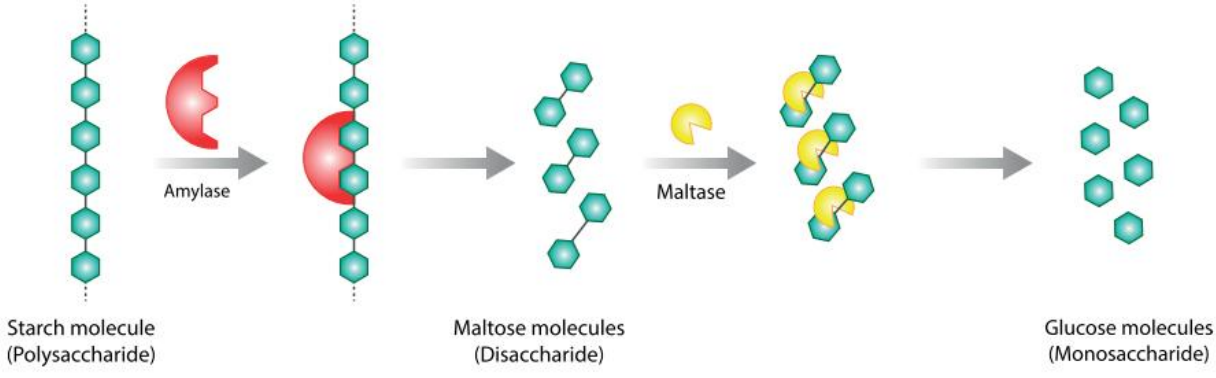


Figure 8.5: Carbohydrate digestion – two step breakdown

There are different forms of disaccharides depending on the source. Each disaccharide has its own enzyme.

- maltose – from malt sugar: made up of two glucose molecules (maltase)
- lactose – milk sugar: made up of glucose and galactose molecules (lactase)
- sucrose – table sugar from sugar cane or sugar beets: made up of glucose and fructose molecules (sucrase)

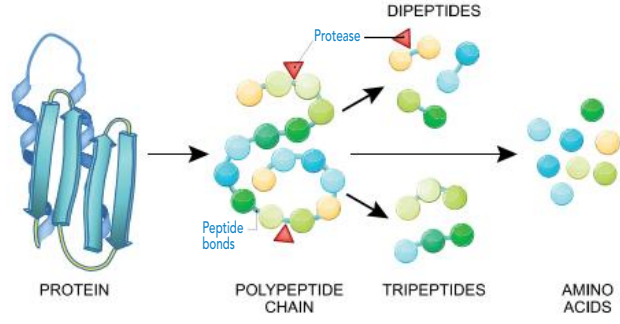


Figure 8.6: Protein digestion – three step breakdown

All the monosaccharides produced can be used in cellular respiration.

Table 8.1: Enzymes, their actions, production locations and action locations

Enzyme	Action	Location of production	Location of action
Amylase	Starch to disaccharides	Salivary glands Pancreas	Mouth Small intestine
Maltase, sucrase, lactase	Disaccharides to monosaccharides	Lining of the small intestine	Small intestine
Lipase	Lipids to glycerol and fatty acids	Pancreas Lining of the small intestine	Small intestine
Protease - pepsin - trypsin - intestinal protease	 Proteins to polypeptide chains Polypeptides to dipeptides Dipeptides to amino acids	 Lining of the stomach Pancreas Lining of the small intestine	 Stomach Small intestine Small intestine
Lipase	Lipids to fatty acids and glycerol	Pancreas Lining of the small intestine	Small intestine

Question 1

State what is meant by optimum conditions for enzymes to function. (1 mark)

Question 2

Describe an active site on an enzyme. (1 mark)

Question 3

State three advantages of using enzymes to catalyse a reaction. (3 marks)

Question 4

Describe the role of co-enzymes. (3 marks)

Question 5

People with haemophilia lack a particular enzyme involved in the chain of reactions required for blood clotting. Explain the results of the lack of this enzyme. (2 marks)

Question 6

Explain why reactions occur at faster rates with increased temperature until a maximum is reached. (6 marks)

Question 7

Outline how the presence of other chemicals can affect the efficiency of an enzyme. (10 marks)

Question 8

Describe how the digestive tract changes to help with the efficient functioning of digestive enzymes. (2 marks)

Question 9

Explain why there are many more proteases than there are lipases in the digestive system. (6 marks)

Question 10

Explain how bile improves the effectiveness of lipases in the small intestine. (4 marks)

LIVER

9

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The salivary glands, pancreas, liver and gall bladder produce or store secretions which aid the processes of digestion.				

The liver is the largest organ of the body. It is divided into sections called lobes.

The liver is an essential organ of the body that performs many vital functions.

Functions of the liver include:

- the deamination of unwanted or excess amino acids to produce urea – a nitrogen containing metabolic waste
- the production of bile which emulsifies fats. Bile is stored in the gall bladder
- the detoxification of poisonous substances such as alcohol and drugs
- the breakdown of hormones
- it stores excess blood glucose as glycogen
- it processes the breakdown products from red blood cells and stores iron for the production of new erythrocytes
- it stores vitamins such as Vitamins A and D until needed by the body cells
- it makes plasma proteins, including those required for blood clotting
- it produces heat through the large number and rate of reactions occurring in cells
- it produces and stores cholesterol that is required for the production of cell membranes
- the metabolism of fats and storage of fats such as triglycerides.

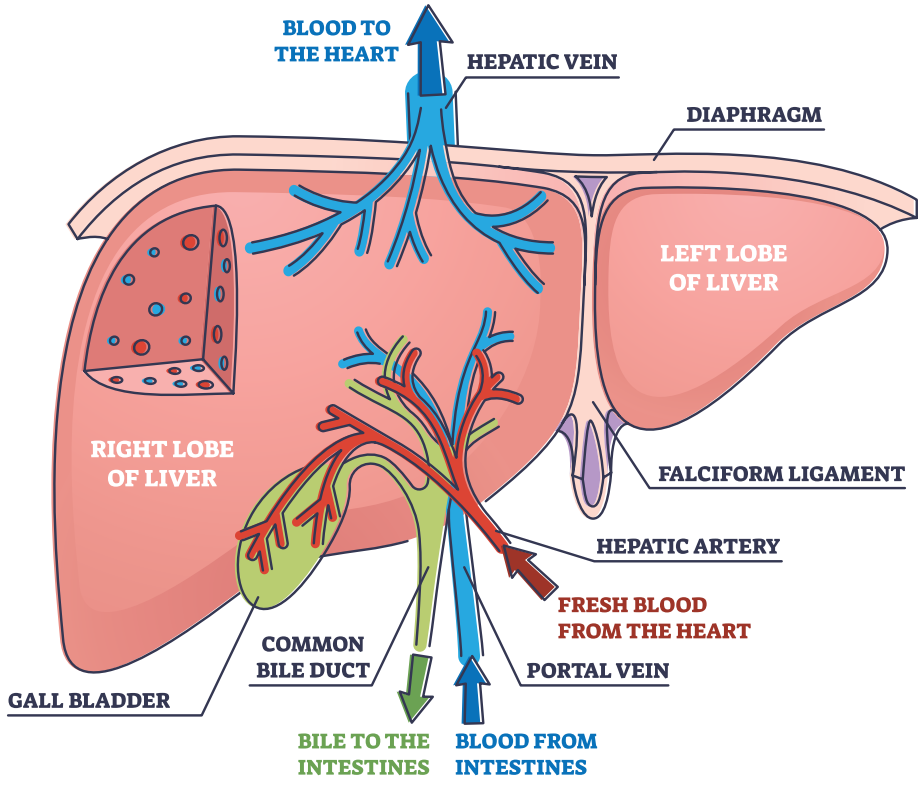


Figure 9.1: Structure of the liver showing the blood flow

Hepatic – relating to the liver, from Latin hepaticus

Hepatitis – inflammation of the liver

The liver receives blood from the intestines via the vein. This branches into venules and capillaries to reach all cells of the liver. It also receives blood from the aorta, again, branching out to all liver lobules. The blood from both sources combine and are returned to the heart via the hepatic vein. This is the only case in the body where blood flows through two capillary beds (one in the intestine to absorb nutrients and the other to deliver nutrients to the liver cells) before returning to the heart.

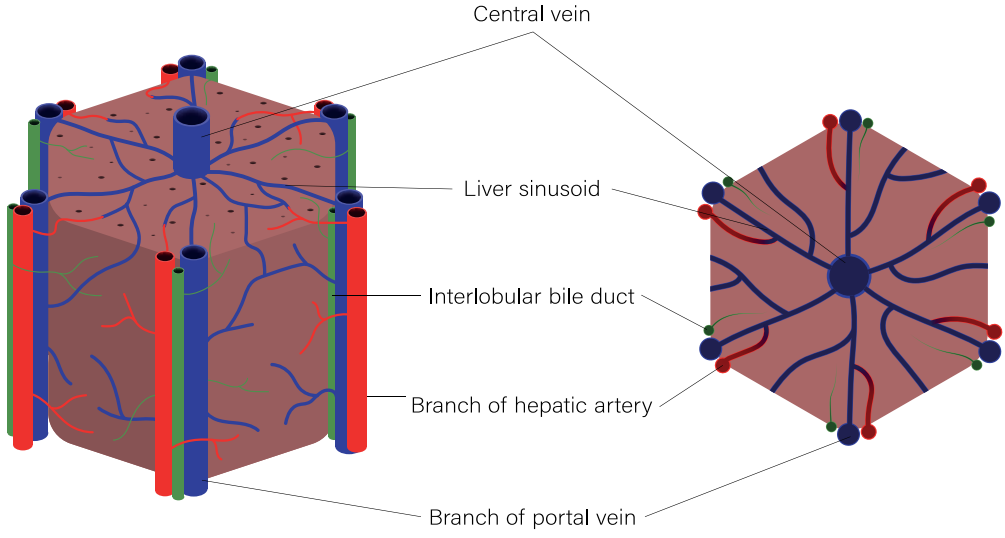


Figure 9.2: Blood flow through the liver lobules

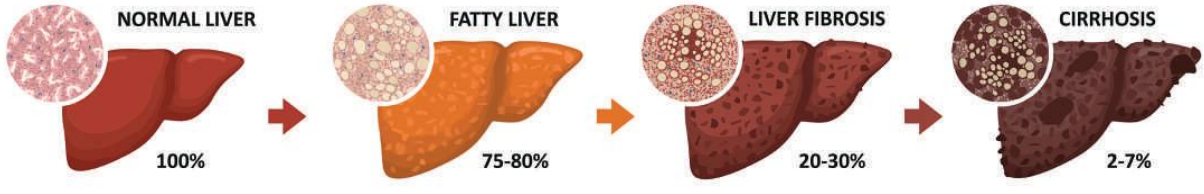


Figure 9.3: Stages of liver damage, showing the percentage of normal function at each stage

Lifestyle choices can affect the liver, causing it to malfunction or reduce its efficiency. Liver disease doesn't always cause noticeable signs and symptoms. If signs and symptoms of liver disease do occur, they may include:

- skin and eyes that appear yellowish (jaundice)
- abdominal pain and swelling
- swelling in the legs and ankles
- itchy skin
- dark coloured urine
- pale coloured faeces
- chronic fatigue
- loss of appetite
- tendency to bruise easily

These symptoms are related to the functions of the liver.

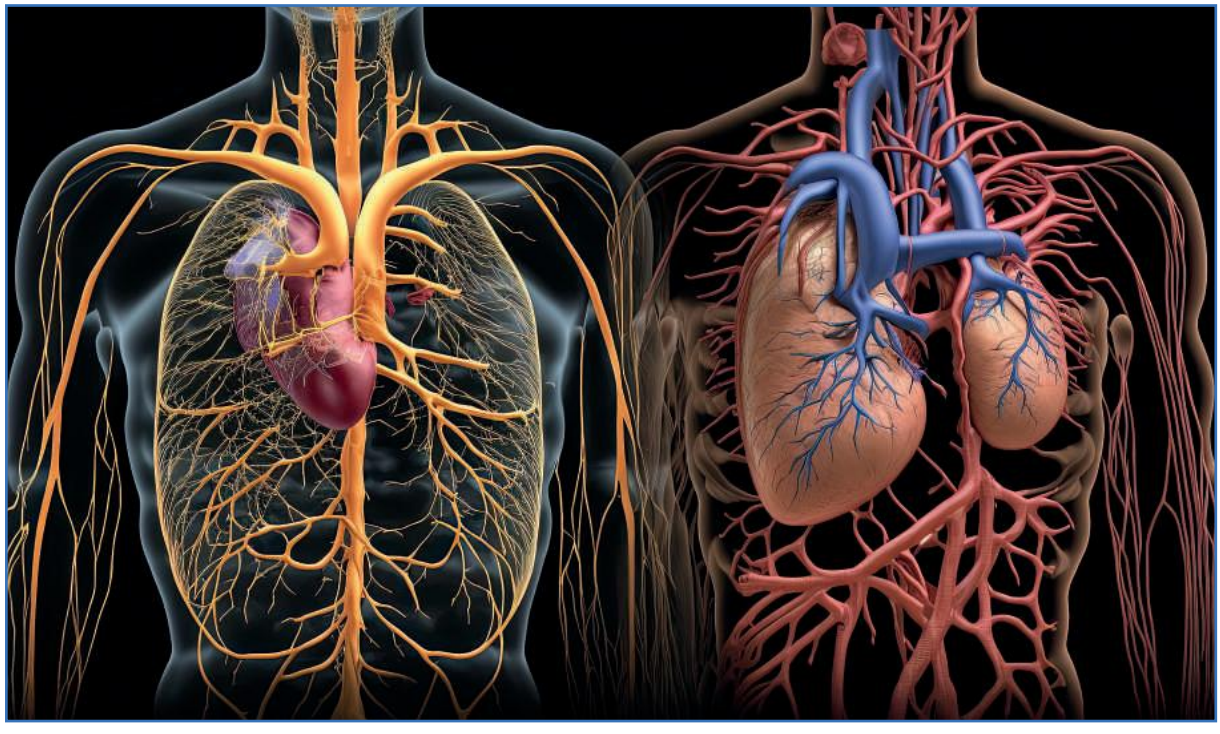
Question 1

Complete the following table with the liver function that changes the contents from the IN to the OUT. (1 mark each)

IN	Liver Function	OUT
high blood sugar		average blood sugar
low blood sugar		average blood sugar
low cholesterol		high cholesterol
high alcohol		lower alcohol
high toxins		low toxins
high amino acids		low amino acids
high triglycerides		low triglycerides
high penicillin		lower penicillin
high thyroid/steroid hormones		low thyroid/steroid hormones
high bilirubin		low bilirubin
high vitamin A, B12, D, E, K		lower vitamin A, B12, D, E, K
high minerals Fe, Cu		low minerals Fe, Cu
worn out erythrocytes, leucocytes		no worn out erythrocytes, leucocytes
inactive vitamin D		active vitamin D (calciferol)
hormones		inactive breakdown products
high bile salts		high volume of bile

CIRCULATORY SYSTEM

10



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The transport of materials within the internal environment for exchange with cells is facilitated by the structure and function of the circulatory system at the cell, tissue and organ levels.				
• Lifestyle choices, including being active or sedentary, the use of drugs and type of diet, can compromise body functioning in the short term and may have long-term consequences.				

The circulatory system (also called the cardiovascular system) connects the external environment with all cells in the body and all cells with all other cells.

Humans have a closed circulatory system in which the flow of blood is contained within blood vessels and is controlled by the pressure from the heart and changing the diameter of the blood vessels.

Functions of the circulatory system linked to other systems

1. Transport of gases – oxygen and carbon dioxide (respiratory system)
2. Transport of nutrients and wastes (digestive and excretory systems)
3. Transport of hormones from endocrine glands to target cells (endocrine system)
4. Regulation of body temperature through the distribution of heat generated from cellular respiration (all body systems by maintaining optimum temperature)
5. Fighting infection through the presence of white blood cells and platelets (immune system)

There are two circuits to the human circulatory system:

- **pulmonary** – from the right side of the heart to the lungs via the pulmonary artery and returns to the left side of the heart via the pulmonary vein
- **systemic** – leaves the left side of the heart to connect with all other parts of the body before returning to the right side of the heart.

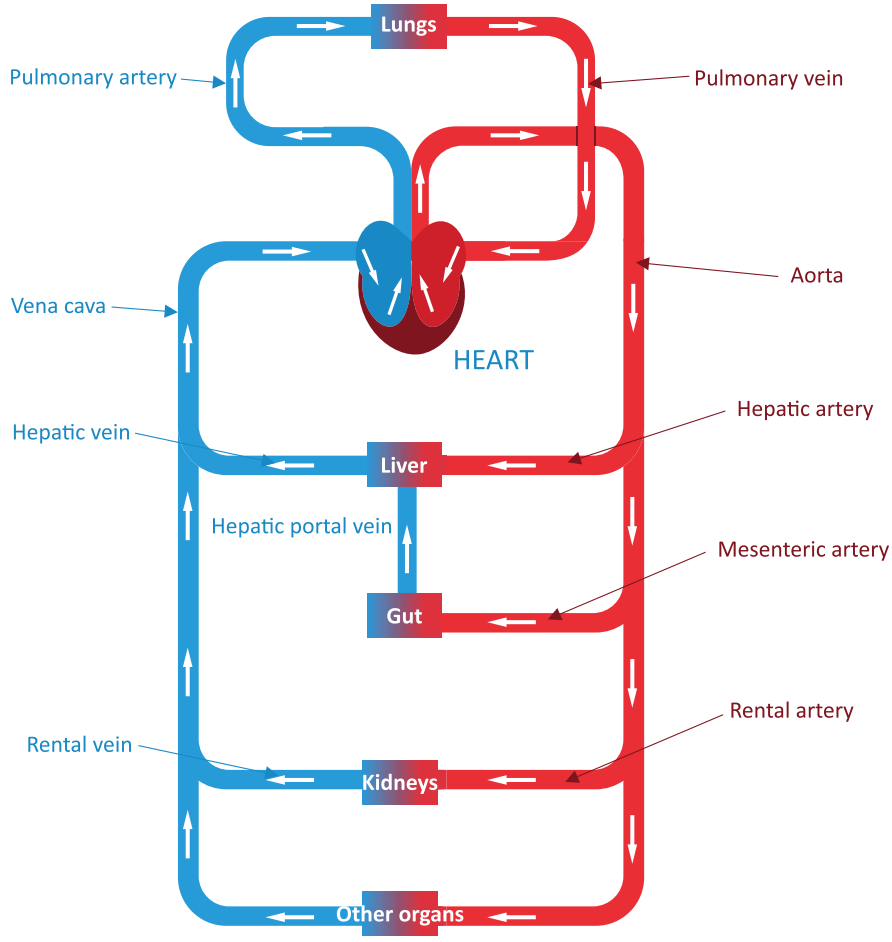


Figure 10.1: Movement of blood in the human circulatory system

HEART STRUCTURE

- four chambers of the heart
- separates pulmonary and systemic circulation
- atria receive blood from the body or lungs
- ventricles pump blood to the body or lungs
- valves control the direction of blood flow in the heart
- veins are attached to the atria
- arteries are attached to the ventricles

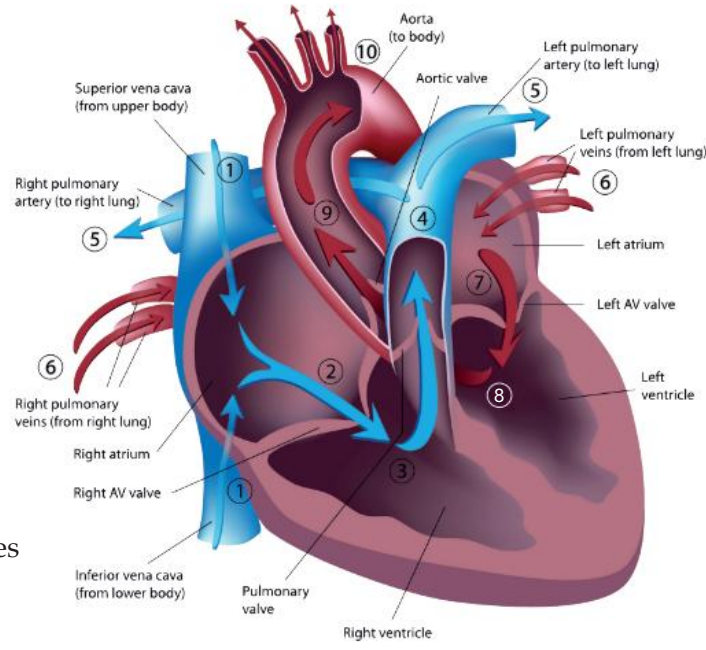
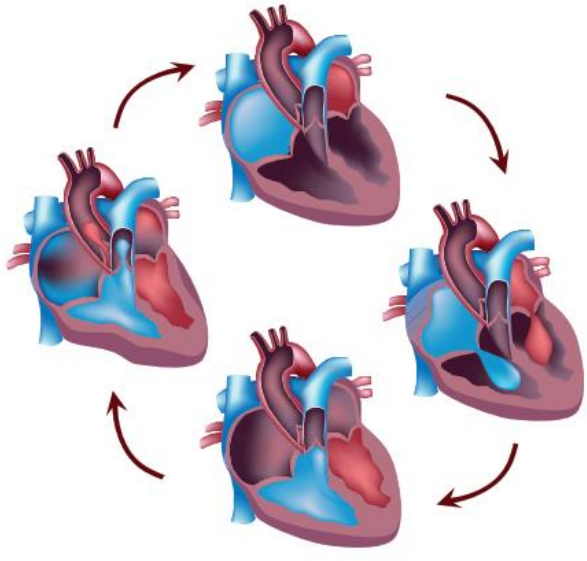


Figure 10.2: Heart structure and pathway of blood through the heart (numbered 1 to 10).

1. Ventricles contract. The left ventricle forces blood through the semi-lunar valves into the aorta.
The right ventricles forces blood through the semilunar valve into the pulmonary artery.



2. The atria relax allowing blood to flow in to the left atrium from the pulmonary veins and into the right atriumfrom the vena cavae.
The tricuspid and bicuspid valves are closed.

3. The ventricles relax, allowing blood to flow in from the atria through the tricuspid (mitral) valve on the right and the bicuspid valve on the left.

4. The ventricles fill and then contract and the cycle starts again.

Figure 10.3: Operation of heart valves

BLOOD VESSELS

Arteries carry blood **away** from the heart. Veins **return** blood to the heart.

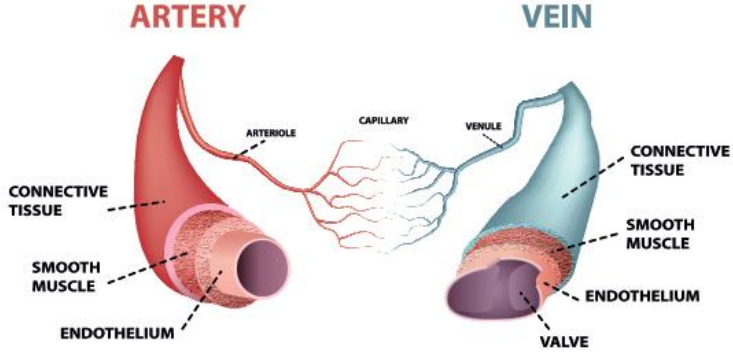


Figure 10.4: Comparison between arteries and veins

Table 10.1: Differences between arteries and veins

Feature	Arteries	Veins
Walls	Highly muscular, thick and contain elastic fibres	Thin with little muscle tissue or elastic fibres
Valves	None	Yes
Colour	Red/pink due to elastic fibres	Dark red
Location	Situated deep in muscles	Found closer to the skin
Oxygen Level	High, except for pulmonary artery	Low, except for pulmonary veins
Nutrient level	High	Low, except for hepatic portal vein between intestines and liver
Blood flow direction	Away from the heart	Towards the heart
Blood pressure	High and decreasing away from the heart	Low and decreasing towards the heart

No exchange of materials happens between arteries and veins and the surrounding tissues.

Arteries and veins are **impermeable**, i.e. materials carried in the blood do not pass through the walls of the arteries or veins into or from the surrounding tissue.

Exchange of materials between blood and the body tissues occurs only at the capillaries.

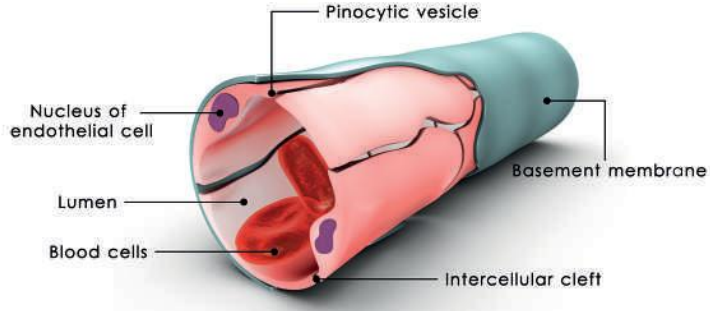


Figure 10.5: Structure of a capillary

Capillary walls are one cell thick with relatively wide spaces between the cells to allow materials to move into and out of the blood.

The surface-area-to-volume ratio of the capillaries is enormous to allow for quick exchange of materials between the blood and the surrounding cells.

All body cells are located within a few cells distance of a capillary. The small distances allow for efficient exchange of materials between the cells and the blood.

Blood flow in capillaries is slow compared to that in arteries and veins and this allows time for the exchange of materials.

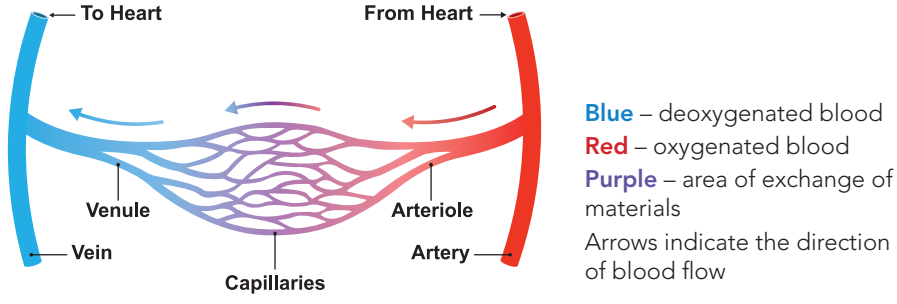


Figure 10.6: Blood flow between blood vessels.

BLOOD PRESSURE

- Blood pressure changes throughout the day depending on many factors including your general health and activity levels.
- Blood pressure can be measured manually, with a blood pressure cuff and stethoscope, or with an automated blood pressure machine called a **sphygmomanometer**.
- The standard units if blood pressure are mmHg which indicates the height of the column of mercury the blood pressure can support. This is based on the original sphygmomanometers which used a column of mercury in a standard sized tube the measure blood pressure. kPa (kilopascals) is another measure of pressure, but the most widely and commonly used is mmHg.

When blood is pumped by the heart around the body, the pressure of the blood pushing against the walls of blood vessels changes.

Blood pressure readings are made up of two values:

- **Systolic blood pressure** is the pressure when the heart muscle is contracting (squeezing) and pumping blood into the arteries from the ventricles.
- **Diastolic blood pressure** is the pressure on the blood vessels when the heart muscle relaxes and blood flows from the atria into the ventricles. The diastolic pressure is always lower than the systolic pressure.

Normal blood pressure – systolic under 140 mmHg and diastolic under 90 mmHg

High blood pressure – systolic over 140 mmHg and/or diastolic over 90 mmHg

High blood pressure is referred to as **hypertension**.

Various conditions and behaviours make high blood pressure more likely. Known as risk factors include:

- leading a sedentary lifestyle (a lifestyle with little or no exercise)
- smoking
- being obese
- a diet with a high salt intake and/or high alcohol consumption
- high blood cholesterol level
- a family history of high blood pressure
- diabetes

Hypertension increases your chances of developing cardiovascular disease including heart attack and stroke, chronic kidney disease, eye disease and erectile dysfunction.

CARDIO-VASCULAR DISEASE

Cardiovascular diseases (CVDs) are a group of disorders of the heart and blood vessels including:

- Coronary heart disease – disease of the blood vessels supplying the heart muscle
- Cerebrovascular disease – disease of the blood vessels supplying the brain
- Peripheral arterial disease – disease of blood vessels supplying the arms and legs
- Rheumatic heart disease – damage to the heart muscle and heart valves from rheumatic fever, caused by streptococcal bacteria
- Congenital heart disease – malformations of heart structure existing at birth.
- Deep vein thrombosis and pulmonary embolism – blood clots in the leg veins, which can dislodge and move to the heart and lungs.

Figure 10.7 shows the deaths due to cardiovascular diseases in 2021.

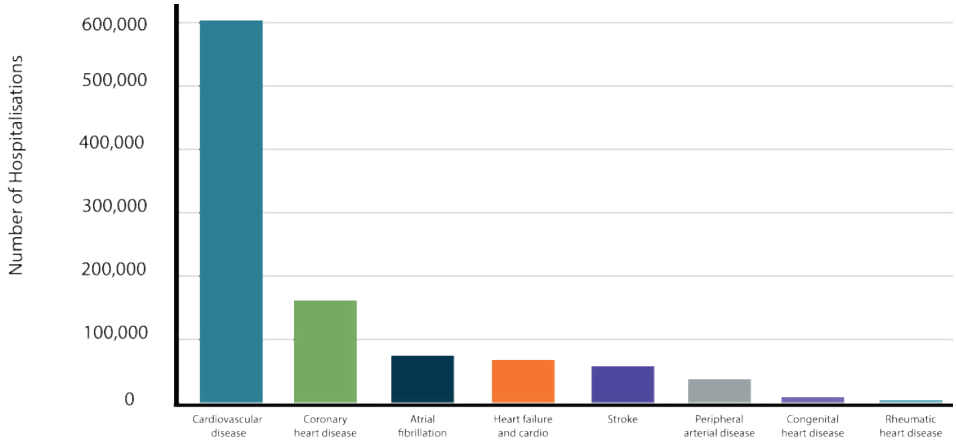


Figure 10.7: Cardiovascular disease deaths in Australia in 2021
From: https://www.aihw.gov.au/reports/heart-stroke-vascular-diseases/hsvd-facts/contents/data-visualisations

Heart attacks and **strokes** are usually acute events and are mainly caused by a blockage that prevents blood flowing from the heart muscle or brain. The most common reason for this is a build-up of fatty deposits on the inner walls of the blood vessels that supply the heart or brain. Strokes can also be caused by bleeding from a blood vessel in the brain or from blood clots.

The most common cause of CVD is the gradual clogging of blood vessels by fatty or fibrous material. Fatty material gradually builds up on the blood vessel walls, narrowing the arteries. This eventually prevents vital oxygen from reaching the cells. As the deposits build up the arteries become less elastic. This condition is often referred to as **hardening of the arteries**. Any artery in the body can be affected. However, the arteries to the heart, brain or kidneys, or those to the eyes and legs are most commonly affected.

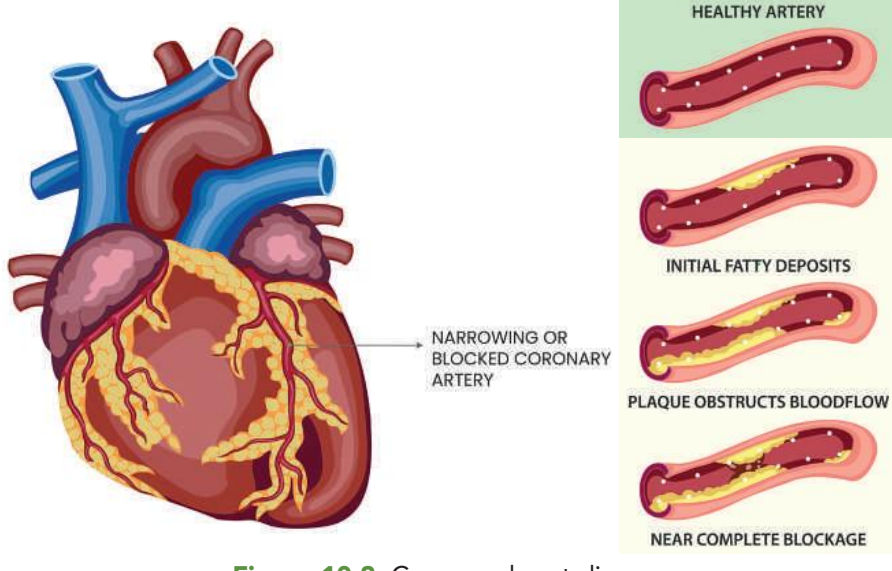


Figure 10.8: Coronary heart disease

Certain groups in the population have significantly higher mortality from heart, stroke and vascular disease than other groups, particularly Indigenous Australians and those of lower socio-economic status.

It is now agreed that most of the **premature deaths** and much of the morbidity caused by heart, stroke and vascular disease are preventable.

This suggests that prevention can occur on a broad front and bring even wider gains than those relating only to **cardiovascular health**.

Main risk factors for developing cardiovascular disease are:

High Blood Pressure

High blood pressure (or hypertension) is a common condition of the circulatory system and is widely recognised as the leading risk factor for CVD.

One third of Australian adults have high blood pressure with more males than females with hypertension.

High Cholesterol

High blood cholesterol is a significant risk factor for developing CVD. Cholesterol is a fat-like substance necessary to make hormones and vitamin D, and to help digest food. The body produces cholesterol, and it's also in some foods.

More than two in five Australian adults are living with high cholesterol.

Being Overweight or Obese

Overweight is identified as having a body mass index (BMI) of 25 or over.

Two in three Australian adults are overweight or obese, with the prevalence of overweight and obesity increasing with age and males more likely to be overweight than females.

Smoking

The smoking statistics are for current smokers, defined as a person who smokes daily, or more than weekly. Vaping is now being included in the statistics collected in this category.

Physical Inactivity

Physical inactivity refers to not getting the recommended level of regular physical activity.

Four in every five adults do not meet national physical activity guidelines, with females being slightly more likely than males to fail to meet physical activity guidelines compared to males. These guidelines are available at *https://www.health.gov.au/topics/physical-activity-and-exercise/physical-activity-and-exercise-guidelines-for-all-australians*

Alcohol Consumption

The Australian guidelines recommend healthy adults should drink no more than ten standard drinks a week, or a maximum of four standard drinks on any one day to reduce the risk of alcohol-related disease.

Males aged 55 to 64 years and females aged 35-44 are most likely to exceed alcohol guidelines.

These guidelines can be found at *https://fare.org.au/wp-content/uploads/FARE-Fact-Sheet-Alcohol-guidelines.pdf#:~:text=The%20Australian%20Alcohol%20Guidelines%20advise%20that%20healthy%20adults,their%20risk%20of%20an%20injury%20from%20alcohol%20use."FARE-Fact-Sheet-Alcohol-guidelines.pdf*

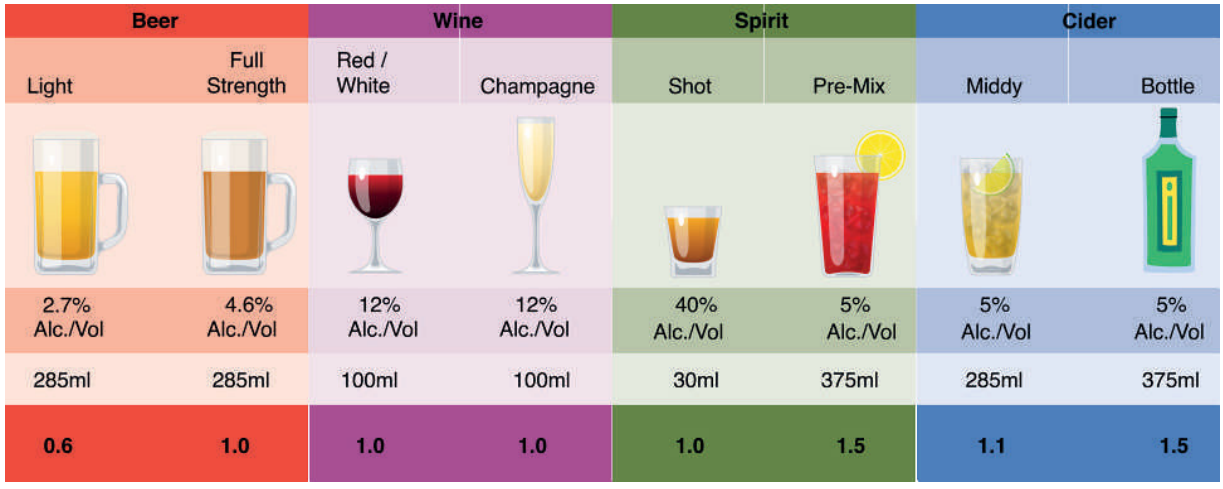


Figure 10.9: Standard drinks in common alcoholic beverages

Diet

Unhealthy diet is one of the leading risk factors for heart disease in Australia. Australians of all ages generally do not eat enough of the five recommended food groups and eat too many junk foods high in salt, fat and sugar.

Ninety-two percent of Australian adults do not meet the recommended intake for vegetables (5+ servings of vegetables a day). Improving vegetable intake to meet the recommended five serves per day is estimated to reduce the risk of cardiovascular disease (CVD) by 16 percent.

From: *https://www.heartfoundation.org.au/bundles/for-professionalsation*

Task 10.1: Complete the following table to assess your CVD risks. (You may like to do a risk assessment for your parents, grandparent or others) Use a scale of 1–5 with 1 being no risk and 5 being high risk.

Risk factor	High BP	High Cholesterol	Weight	Smoking (including vaping)	Alcohol	Diet
My risk						

Question 1

List the materials exchange in the purple zone in Figure 10.6. (9 marks)

Moving out of the blood	Moving into the blood

Question 2

Explain how the structure of the heart helps with the efficiency of exchanges of materials. (5 marks)

Question 3

Some children are born with a 'hole in the heart'. This hole is usually located in the wall between the left and right ventricles. Explain how this would affect the supply of oxygen to cells. (6 marks)

Question 4

Some people may have a 'heart murmur' which could indicate the malfunction of one of the heart's valves. Outline the problems that could arise from having a heart murmur. (3 marks)

Question 5

Explain why the aorta has very thick elastic walls compared to any other blood vessels. (4 marks)

Question 6

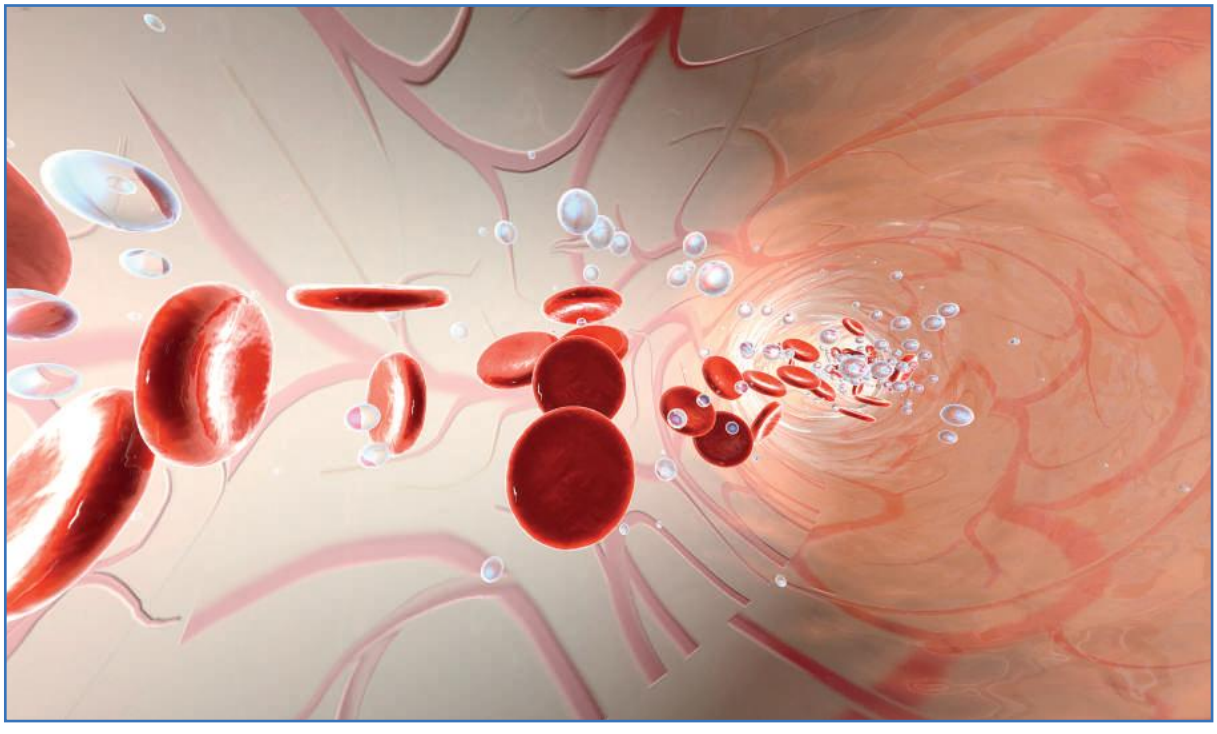
The heart muscle tissue does not get nutrients from the blood in the chambers. Explain why. (4 marks)

Question 7

In the case of a stroke, a blood clot lodges in the fine arterioles leading to a capillary bed. Explain what would happen to the tissues surrounding that capillary bed if the blood clot was not cleared quickly. (3 marks)

BLOOD AND TRANSFUSIONS

11



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The components of blood facilitate the transport of different materials around the body (plasma and erythrocytes), play a role in the clotting of blood (platelets) and the protection of the body (leucocytes).				
• Blood transfusions rely on determining blood groups (ABO and Rhesus factor), and can be used to treat many different diseases and conditions.				

BLOOD CONTENTS

Blood is the fluid that circulates in the cardiovascular system.

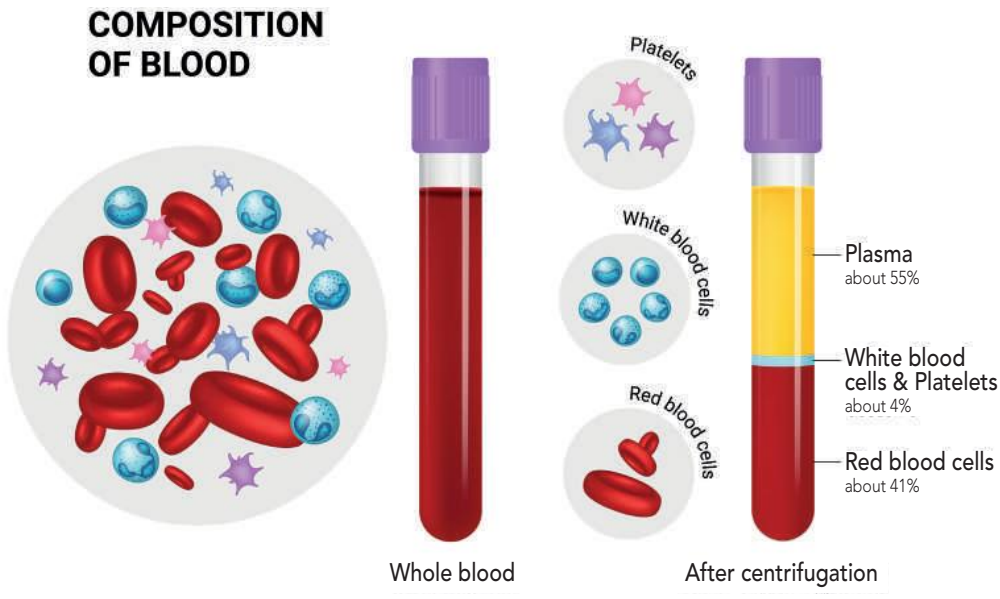
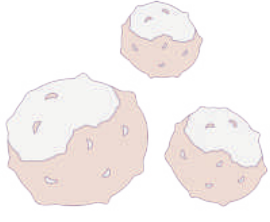
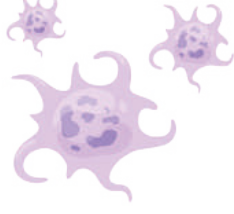


Figure 11.1: Blood contents

Table 11.1: Cellular contents of blood

Cell type	Description	Function
	Erythrocytes, or red blood cells, contain no nucleus last about 120 days before being replaced	Transport oxygen attached to haemoglobin which increases the oxygen-carrying capacity of the blood compared with direct dissolving of oxygen in plasma About 10% of carbon dioxide transport is by attachment to haemoglobin to form carboxyhaemoglobin
	Leucocytes, or white blood cells, have a large nucleus or several nuclei	Responsible for protecting the body against infection by recognising and eliminating pathogens, producing antibodies and cleaning up damaged cells
	Thrombocytes, or platelets, are cell fragments with no nucleus	Recognise damaged blood vessels and prevent and stop bleeding

ERYTHROCYTES

Because erythrocytes, also known as red blood cells, do not have a nucleus, they are unable to produce proteins, repair themselves, grow, or divide.

- **Life span.** As they age, erythrocytes become more rigid and begin to fragment, or fall apart, in **about 120 days.**
- **Haemolysis** or breakdown of erythrocytes occurs in the spleen and liver.
- **Replacement of erythrocytes** is more or less continuous by the division of haemocytoblasts (stem cells) in the red bone marrow.

THROMBOCYTES

Thrombocytes, also known as platelets, are formed when cytoplasmic fragments of megakaryocytes, which are very large cells in the bone marrow, pinch off into the circulation as they age. Platelets play an important role in the formation of a blood clot by:

- gathering to block a damaged blood vessel
- providing a surface on which strands of fibrin protein form an organised clot
- contracting to pull the fibrin strands together to make the clot firm and permanent
- stimulating the activity a series of clotting factors necessary to the formation of the clot.

LEUCOCYTES

Leukocytes, also known as white blood cells, have different forms that perform different roles in the immune system. Some of these are outlined below.

- Neutrophils are the first responders of immune cells.
- Basophils release histamine to mount a non-specific immune response.
- Eosinophils fight bacteria and parasites, but also provoke allergy symptoms.
- Lymphocytes are B and T cells that defend against specific pathogenic invaders.
- Monocytes and phagocytes clean up dead cells.

PLASMA

Plasma is the aqueous part of blood containing proteins and salts in which red and white blood cells and platelets are suspended. It constitutes approximately 55 percent of total blood volume. Plasma has numerous functions including those below.

- Redistributing water to where it is needed in the body.
- Transporting hormones, nutrients and proteins to parts of the body and helping to exchange oxygen and carbon dioxide.
- Supporting blood vessels from collapsing or clogging.
- Regulating body temperature by absorbing and releasing heat.
- Removing waste from cells and transporting it to the liver, lungs and kidneys for excretion.
- Containing factors important in blood clotting.
- Defending against bacterial, viral, fungal and parasitic infections.

BLOOD TRANSFUSIONS

Blood transfusion is when blood from one person is transferred into the circulatory system of another. Blood transfusions are only successful when the blood of the donor matches that of the recipient.

Blood groups are determined by the presence of particular proteins called **antigens**, on the membranes of erythrocytes. The types of antigens present is genetically controlled.

The ABO system is used to classify blood for transfusion into compatible recipients.

Blood is also classified by the presence or absence of Rhesus D factors or Rh factors, another type of protein on the erythrocyte membrane.

By using these two systems together, the success of a transfusion increases.

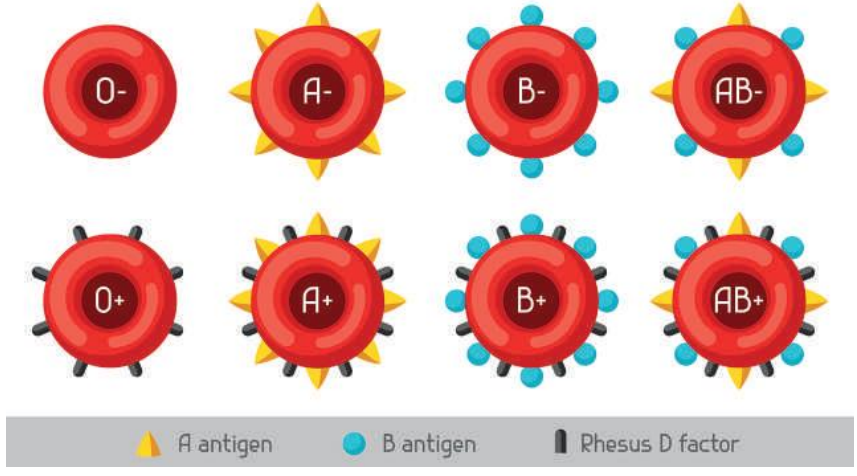


Figure 11.2: Blood types based on proteins present on the erythrocyte membrane

Blood typing is done by adding antibodies to blood samples to observe the results. Clotting or clumping of cells means the antigen is present.

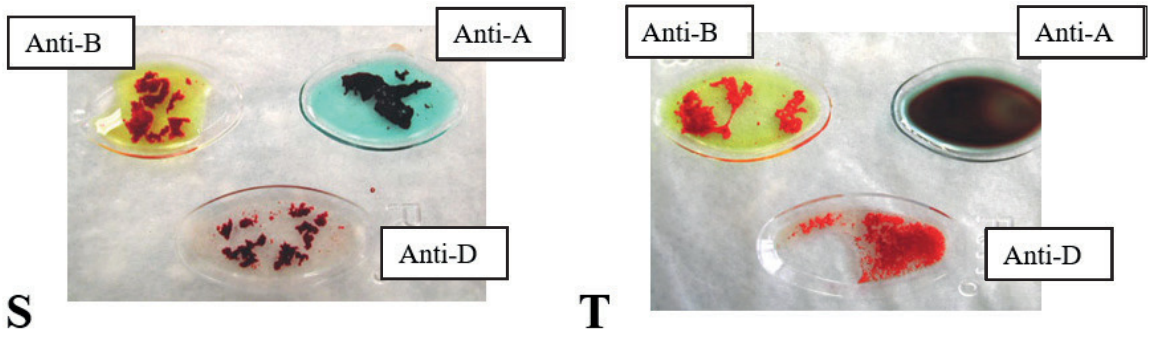


Figure 11.3: Blood type testing showing agglutination

Task 11.1: Determine the blood type of Persons S and T and justify your suggestions.

When the blood type is known, blood donations that can be used for transfusions can be selected. Figure 11.4 shows the blood types that are compatible: note whether the person is a donor or a recipient.

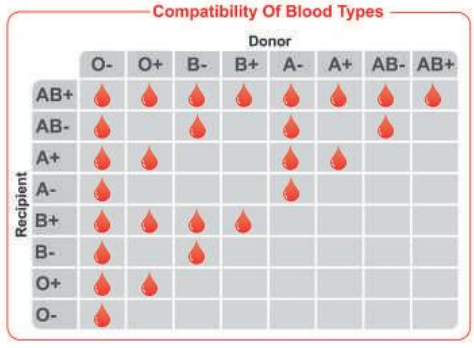


Figure 11.4: Compatibility of blood types for transfusions

Task 11.2: Use Figure 11.4 to determine a person who could be called:

- a universal recipient.
- a universal donor.

BLOOD DONATION

Volunteers can donate blood at Australian Red Cross Lifeblood donor centres.

Donations can be in the form of whole blood, around 500 mL, or:

- plasma – a centrifuge separates the blood contents and collects the plasma. The rest of the contents are returned to the circulation during the donation process.
- platelets – a centrifuge separates the blood components and collects the platelets and some plasma. The rest is returned to the circulation.

USES OF BLOOD PRODUCTS

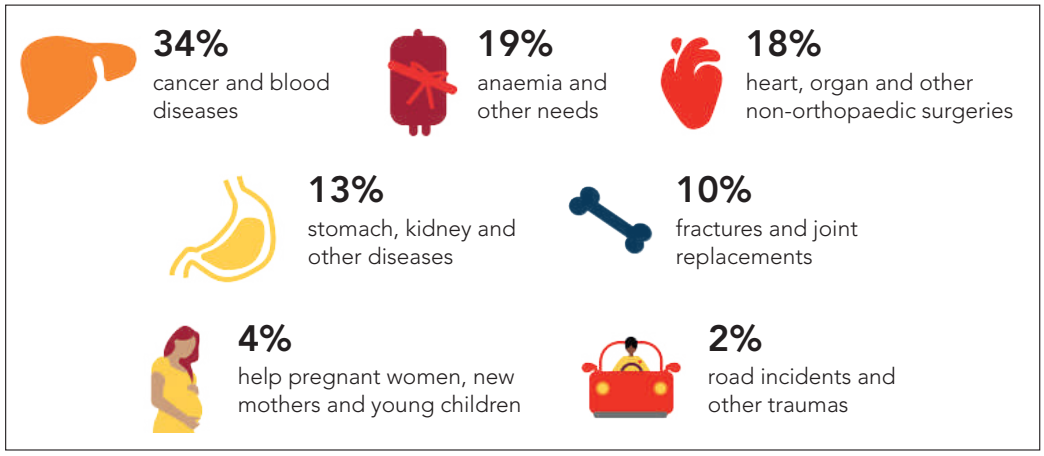


Figure 11.5: Uses of donated red blood cells
From: https://www.lifeblood.com.au/blood/learn-about-blood/why-donate-blood

Table 11.2: Uses of plasma

Condition	Reason
Chicken pox	Chickenpox can be a serious illness. Zoster Immunoglobulin in donated plasma provides temporary protection against chickenpox.
Immune deficiencies	Donated plasma contains antibodies. These plasma can be mixed with plasma from other people to fight an army of pathogens.
Rh disease	In some cases when two people with different Rh (D) types (that's the +/- in your blood type) have a child, an Rh (D) negative mother's immune system may produce antibodies that can destroy the red blood cells of their Rh (D) positive child. Plasma helps prevent Rh disease, with a treatment given to affected pregnant women.
Measles	For those who can't be vaccinated against measles, plasma can save the day. It's used to make normal human immunoglobulin (NHIG), which can provide temporary protection.
Liver disease	Liver damage reduces the production of clotting agents. Plasma can help prevent or stop bleeding complications that arise from liver disease.
Cancerous and non-cancerous diseases	Bone marrow replacement patients are at particular risk of very severe infections. The antibodies within plasma can help prevent infections.
Haemophilia	Haemophilia affects the blood's ability to clot. Plasma can be used to make products that can replace missing clotting factors.
Haemorrhages	A transfusion of a special plasma product that replaces vital clotting factors, can help to stop critical bleeding from trauma or surgery.
Rare blood disorders	For some inherited rare blood disorders often the treatment is quite simple – regular transfusions of treatments made from plasma.
Kidney disease	If the kidneys fail due to kidney disease, albumin, a protein extracted from plasma, can be used in the treatment.
Severe burns	Plasma can offer patients suffering severe burns some relief. Albumin, can help replenish and replace lost fluid and proteins, preventing a patient from going into shock.

PLATELETS

People with leukaemia or going through treatments like chemotherapy can have their platelet counts become so low that they have spontaneous bleeding.

Even a small amount of bleeding is dangerous, but it's especially so if it happens in the brain. Platelets can stop that from happening by clotting to stop the bleed.

Platelets can also stop bleeding during surgery or after major trauma, including incidents from a workplace accident to a shark attack.

Question 1

Provide the most appropriate scientific terms for the following.

(a) The process of blood cells clumping together during blood typing tests. (1 mark)

(b) The blood group that can be used for donations to the most other blood types. (1 mark)

(c) The factor that is tested for by Anti-D antibodies. (1 mark)

(d) The chemical that binds with oxygen and carbon dioxide in erythrocytes. (1 mark)

(e) The blood cell type that makes up about 4% of blood volume. (1 mark)

Question 2

Explain why erythrocytes need to be replaced after about 120 days. (5 marks)

Question 3

Describe the ways in which oxygen is carried in the blood. (2 marks)

Question 4

Explain why a lack of iron reduces the oxygen-carrying capacity of blood. (3 marks)

Question 5

Explain why there are so many different types of leucocytes in blood. (2 marks)

Question 6

Explain the role of blood in maintaining a constant body temperature. (2 marks)

Question 7

Haemophiliacs lack a clotting factor, so damaged blood vessels continue to leak blood. Which type of transfusion would be best given to a haemophiliac who is bleeding? (2 marks)

Question 8

Explain why donors can only donate blood every 3–4 months. (3 marks)

Question 9

Arteries are usually coloured red and veins in blue. This is because the blood in arteries appears to be a brighter red than in veins. State the reason for the difference in blood colour in the different blood vessels. (4 marks)

LYMPHATIC SYSTEM

12

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The lymphatic system functions to return tissue fluid to the circulatory system and to assist in protecting the body from disease.				

The lymphatic system is complementary to the circulatory system. Plasma leaks out of the blood capillaries into the tissue fluid and not all of it returns to the blood circulation. The closed system of lymph vessels between the cells of tissues collects the fluid and returns it to the blood circulation.

- The fluid in the vessels is called **lymph** which is similar to **plasma** but with less protein, no erythrocytes and more lymphocytes.
- Lymph vessels are closed-ended with lymph flowing in **one direction**.
- Control of movement of lymph is by **valves** similar to those in veins.
- The lymph is moved along the lymph vessels by the squeezing **action of surrounding muscles**.
- Lymph is returned to the blood circulation via ducts into the left and right subclavian veins.

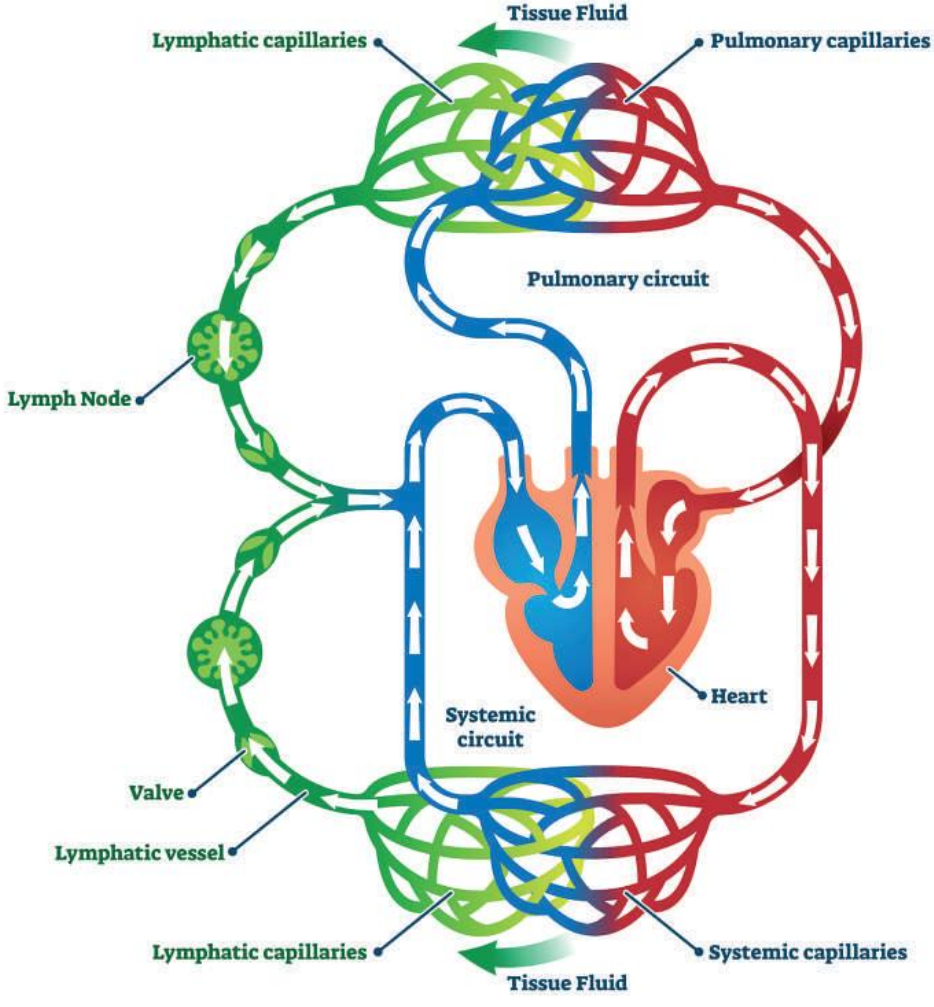


Figure 12.1: Lymphatic circulation alongside blood circulation
Note the direction of the arrows in the lymphatic system vessels.

Key functions of the lymphatic system are:

- maintaining fluid levels in the body tissue by returning excess fluid to the circulation system
- absorbing fats from the digestive tract and transporting them to the blood circulation
- protecting the body against foreign invading pathogens
- transporting and removing waste products and abnormal cells from the lymph.

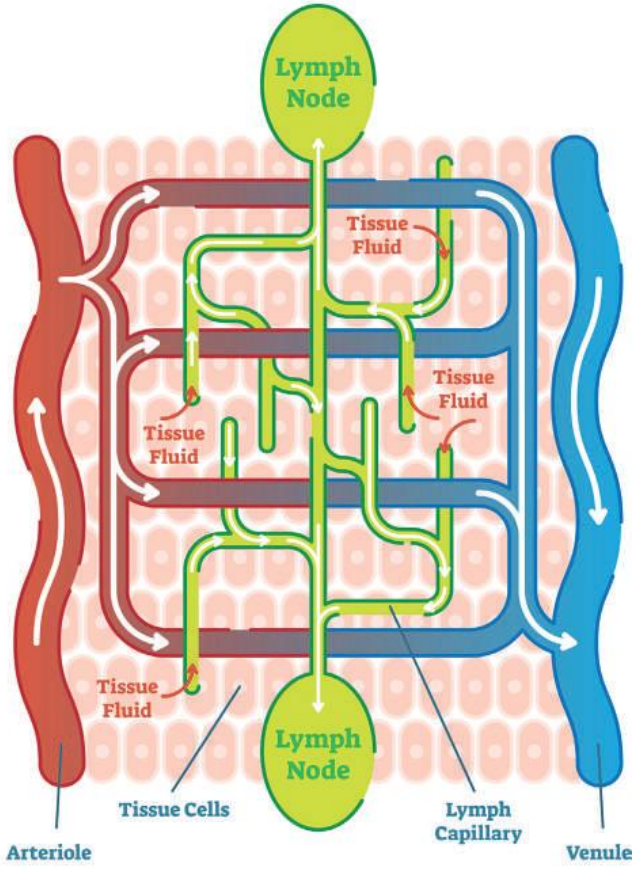


Figure 12.2: Location of lymph vessels and blood vessels in tissue

LYMPH NODES

Lymph nodes are swellings at intervals along the lymph vessels.

- They monitor and cleanse the lymph as it filters through them.
- The nodes filter out damaged cells, pathogens and cancer cells.
- These lymph nodes also store lymphocytes, phagocytes and other immune system cells that attack and destroy bacteria and other harmful substances in the lymph.
- There are about 600 lymph nodes scattered throughout the body. Some exist as a single node, others are closely connected groups called chains.
- A few of the more familiar locations of lymph nodes are in your armpit, groin and neck.

There are other major lymph nodes.

- **Spleen** – this largest lymphatic organ is located on the left side under the ribs and above the stomach. The spleen filters and stores blood and produces white blood cells that fight infection or disease.
- **Thymus** – this organ is located in the upper chest beneath the sternum. It matures a specific type of white blood cell that attack pathogens.
- **Tonsils and adenoids** – these lymphoid organs trap pathogens from the food eaten and the air breathed. They are the body's first line of defence against foreign invaders.
- **Peyer's patches** – these are small masses of lymphatic tissue in the mucous membrane that lines the small intestine. These lymphoid cells monitor and destroy bacteria in the intestines.
- **Appendix** – the appendix contains lymphoid tissue that can destroy bacteria before it breaches the intestine wall during absorption.

Question 1

Many children have bouts of tonsilitis. Explain how this indicates a functioning lymphatics system. (3 marks)

Question 2

Figure 12.3 shows the changes in the shape of a leg of a person suffering from lymphoedema.

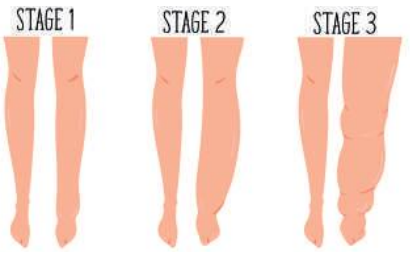


Figure 12.3: Stages of development of lymphoedema

Explain how this change could occur. (2 marks)

Question 3

Compare lymph to blood plasma. (10 marks)

Question 4

When a woman has a radical mastectomy (breast removal) the lymph nodes are commonly removed from the chest, armpit and upper arm on the same side from which the cancerous breast was removed. Explain why this is done. (4 marks)

Question 5

Phagocytes are found in large numbers in the lymph nodes. Described their role. (2 marks)

RESPIRATORY SYSTEM

13

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The exchange of gases between the internal and external environments of the body is facilitated by the structure and function of the respiratory system at the cell, tissue and organ levels.				
• The efficient exchange of gases in the lungs is maintained by the actions of breathing, blood flow and the structure of the alveoli.				
• The treatment of conditions due to system or organ dysfunction has changed through improvements in early diagnosis and appropriate use of drugs, physiotherapy, and removal and/or replacement of affected parts.				
• Lifestyle choices, including being active or sedentary, the use of drugs and type of diet, can compromise body functioning in the short term and may have long-term consequences.				

All living cells must carry out respiration to release energy for life functions.

Living cells need to be supplied with oxygen and have carbon dioxide removed.

With no direct contact with the external environment, humans need to have systems in place to provide this gas exchange service to all cells.

The respiratory system takes air from the environment to be in close contact with the blood for gas exchange of gases to take place.

There are three very distinct processes in supplying cells with their gaseous needs:

- **Breathing** – movement of air into and out of the lungs.
- **Gas exchange** – movement of gases across the membranes of the alveoli, capillaries and cell membranes.
- **Respiration** – chemical reaction: glucose + oxygen → carbon dioxide + water + ATP.

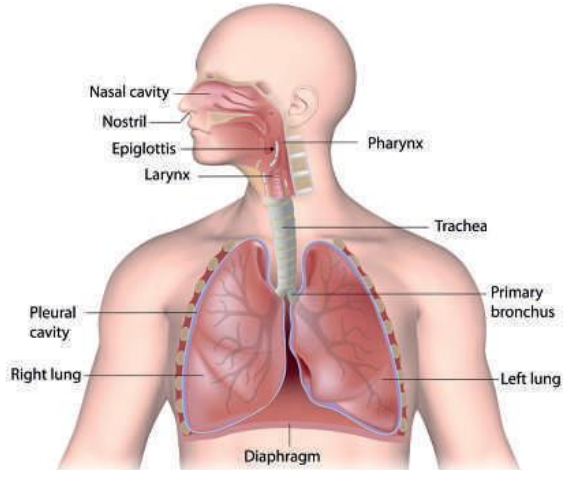


Figure 13.1: The respiratory system

NASAL CAVITY

- Hairs inside the cavity filter dust particles and air-borne pathogens (e.g. fungal spores) from the air.
- Ciliated mucous epithelium that lines the cavity secretes mucus to trap fine dust and bacteria and move them by the cilia to the back of the nose.
- Blood capillaries in the lining of the cavity moisten and warm the air.
- The soft palate closes this off during swallowing to prevent food entering the nose.

PHARYNX

The pharynx is the common area between the respiratory and digestive systems.

EPIGLOTTIS

The epiglottis closes the opening of the trachea to prevent food from getting into the trachea.

LARYNX

The larynx, or voice box, is made of cartilage with epithelial tissue (vocal cords) stretched across the upper end. These cords vibrate during controlled expiration to form the sounds of the voice.

TRACHEA AND BRONCHI

- The trachea and bronchi are flexible tubes passing from the larynx and branching into each lung.
- They are supported by incomplete rings of cartilage.
- The inner surface is lined with ciliated epithelium which secretes mucus to trap particles, including bacteria, and moves them to the pharynx to be swallowed.

LUNGS

- The left lung has two lobes; the right lung has three lobes.
- They are surrounded by pleural membranes as is the inner surface of the thoracic cavity. Between these membranes is pleural fluid which helps reduce friction caused by the movement of the lungs during breathing.
- The bronchi of each lung divides into bronchioles which end in tiny air sacs called alveoli (single: alveolus).

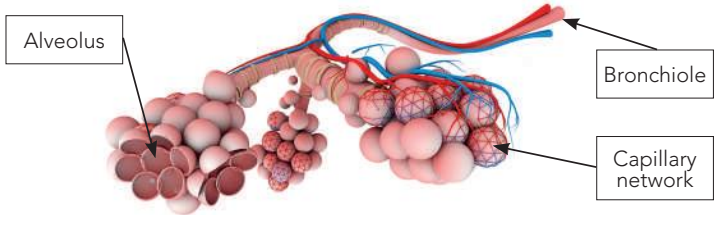


Figure 13.2: The alveoli at the terminals of the bronchioles

ALVEOLI

The alveoli are structured for the efficient exchange of gases by diffusion.

For the **diffusion rate to be efficient**, there needs to be:

- a **large surface-area-to-volume** – lots of small spherical air sacs
- **moist surfaces** – provided by the mucus covering; only dissolved gases will diffuse across the membranes
- **thin membranes** – short distance between the air in the alveolus and the blood in the capillary network to allow for quick movement of gases
- **high concentration gradient** – maintained by the breathing process and the continual movement of blood through the capillary network.

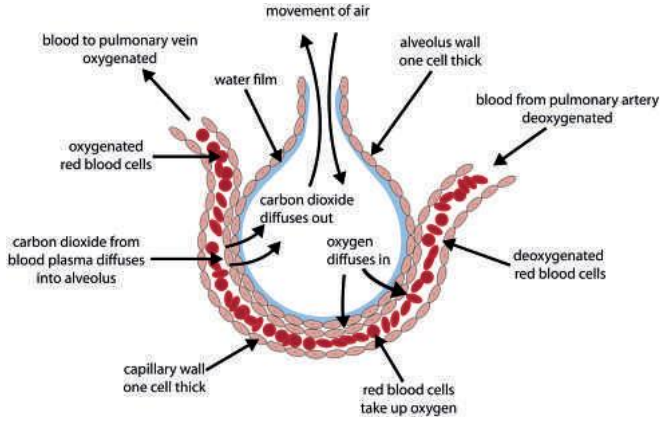


Figure 13.3: The movement of materials at the alveolus for the efficient exchange of gases

BREATHING

Breathing is the movement of the body causing air to move into and out of the lungs. The movement of air is brought about by changing the air pressure between the air in the lungs and the outside. Air will move from an area of high pressure to an area of low pressure.

- **Inspiration**: Air pressure is **reduced** in the lungs by making the volume larger. The contraction and downward movement of the diaphragm and the upward movement of the rib cage increases the chest volume while decreasing the pressure of the air inside the lungs causing the air to move in.
- **Expiration**: Air pressure is **increased** in the lungs by making the volume smaller. The diaphragm relaxes and moves upwards and the rib cage moves downwards reducing the chest volume and increasing the pressure of the air inside the lungs. Air moves out.

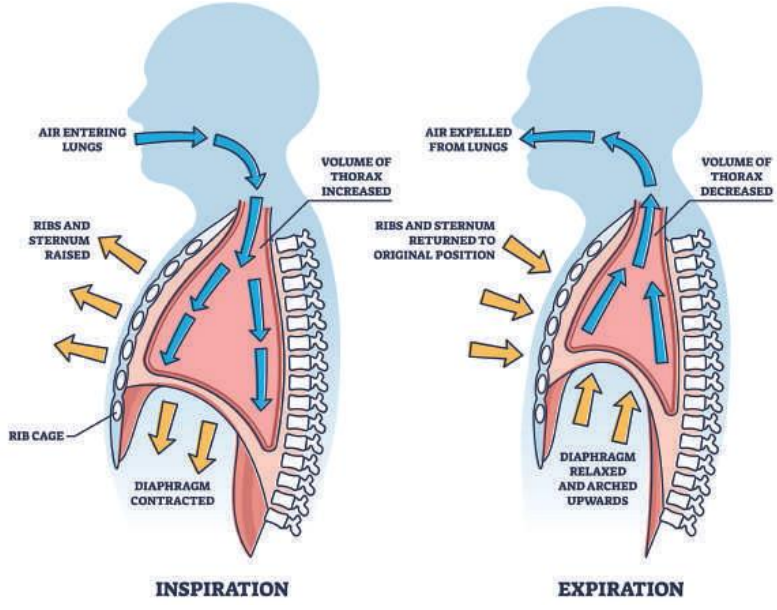
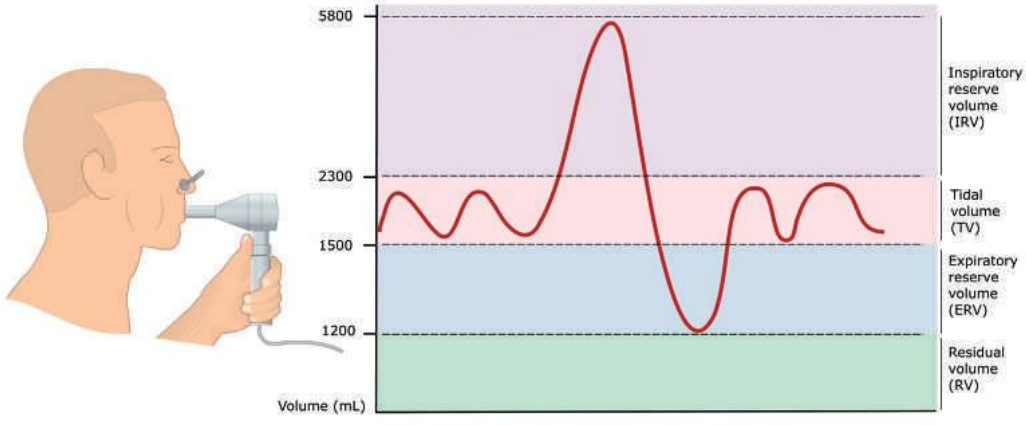


Figure 13.4: Mechanisms of breathing

LUNG CAPACITIES



Male	Female
3000	1900
500	500
1100	700
1200	1100
5800 mL	4200 mL

Figure 13.5: Measuring lung capacities

A spirometer is used to measure volumes of air moving into and out of the lungs.

- Tidal volume – amount of air breathed or out per normal breath
- Inspiratory reserve volume – amount of air forcibly inspired in addition to tidal volume
- Expiratory reserve volume – amount of air forcibly expired in addition to tidal volume
- Vital capacity – maximal amount of air inspired after a maximal expiration (TV + IRV + ERV)
- Residual volume – amount of air left in the lungs after maximal expiration
- Total lung capacity – TV + IRV + ERV + RV

Changes in these volumes can indicate changes in health.

PROBLEMS CAUSED BY SMOKING AND VAPING

Lifestyle choices including smoking and vaping compromise the functioning of the lungs in the short term and can have long term consequences. Toxins from smoking and vaping enter the body via the respiratory system.

- **Cancer**

  Smoking causes most lung cancers and can cause cancer almost anywhere on the body. This includes the lips, tongue, mouth, nose, oesophagus, throat, voice box, stomach, liver, kidney, pancreas, bladder, blood, cervix, vulva, penis and anus.

- **Breathing problems and chronic respiratory conditions**

  Smoking is the main cause of chronic obstructive pulmonary disease (COPD), a serious, progressive and disabling condition that limits airflow in the lungs. Active smoking also worsens asthma and is associated with an increased risk for asthma in adolescents and adults.

- **Heart disease, stroke and blood circulation problems**

  Smoking is major cause of cardiovascular disease, such as heart disease and stroke. Smoking increases the risk of blood clots, which block blood flow to the heart, brain or legs. Some smokers have their limbs amputated due to blood circulation problems caused by smoking.

- **Diabetes**

  Smoking causes type 2 diabetes, with the risk of developing diabetes 30 to 40% higher for active smokers than non-smokers. Smoking may also worsen some of the health conditions related to type 1 diabetes, such as kidney disease.

EMPHYSEMA

Emphysema occurs when the walls of the alveoli are damaged. This causes the small airways in the lungs to collapse when you breathe out. This makes it hard for air to flow into and even harder for it to flow out of your lungs as they stick together due to the presence of the mucous lining of these tubes.

The damage also reduces the wall space making the alveoli become bigger, reducing the surface-area-to-volume ratio. Over time, the lung tissue becomes less elastic and more stiff, reducing the efficiency of gas exchange to levels insufficient to supply the needs of the body cells.

PNEUMONIA

Pneumonia is an infection of the lungs causing breathing difficulties. The alveoli may fill with fluid or pus. This reduces the efficiency of gas exchange because the mucus reduces the surface-area-to-volume ratio and increases the distance the gases need to move between the air and the blood.

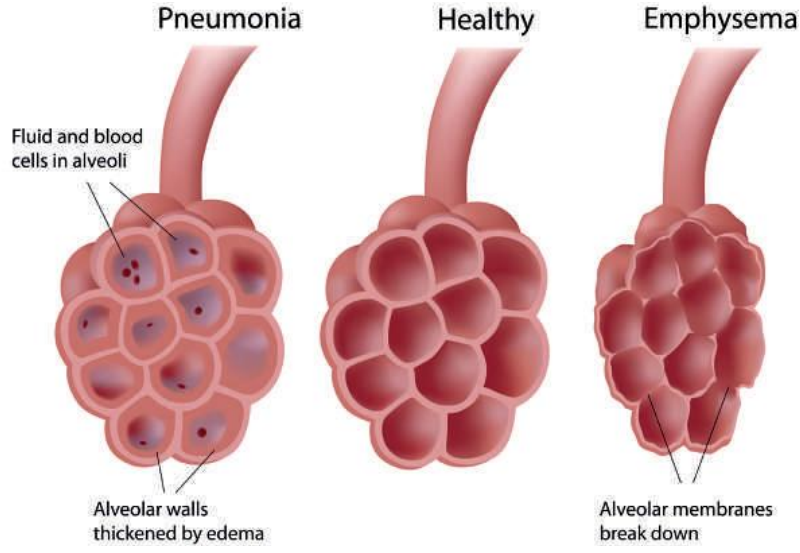


Figure 13.6: Comparison of healthy alveoli with those of pneumonia and emphysema patients

VAPING

The e-cigarette aerosol that users breathe from vaping devices can contain harmful and potentially harmful substances similar to those in cigarettes.

The effects of vaping are very similar to smoking, due to the similar contents, but there is not as much evidence available from scientific research, as the introduction of vaping is recent.

ASTHMA

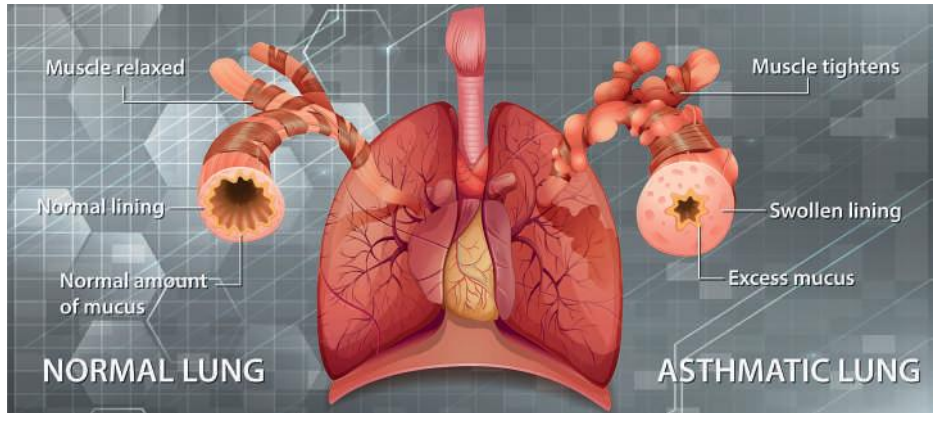


Figure 13.7: Differences between normal airways and those effected by asthma

Asthma is a long-term lung condition that is caused by narrowing of the airways when they become inflamed. Narrowing of the airways reduces the flow of air into and out of the lungs affecting the rate of gas exchange and therefore, metabolic activity. Asthma is caused by particulate matter in the air causing an inflammatory reaction in the lining of the airways.

Medications for asthma relax the muscles surrounding the airways.

Question 1

State the pathway of an oxygen molecule during inspiration. (8 marks)

Question 2

Explain the changes in the movement of the rib cage and the diaphragm during a cough as compared to a normal expiration. (5 marks)

Question 3

Explain how the efficiency of the respiratory system is compromised when a person has a cold. (5 marks)

Question 4

Describe what happens when a person chokes when swallowing food. (4 marks)

Question 5

Explain why it is impossible to talk while swallowing. (3 marks)

Question 6

Outline how tidal volume would change when a person is doing vigorous exercise. (4 marks)

Question 7

Explain how the number of erythrocytes in the blood helps in the diffusion rate of oxygen from the air into the blood. (5 marks)

Question 8

Explain how you can breathe without moving your rib cage. (3 marks)

Question 9

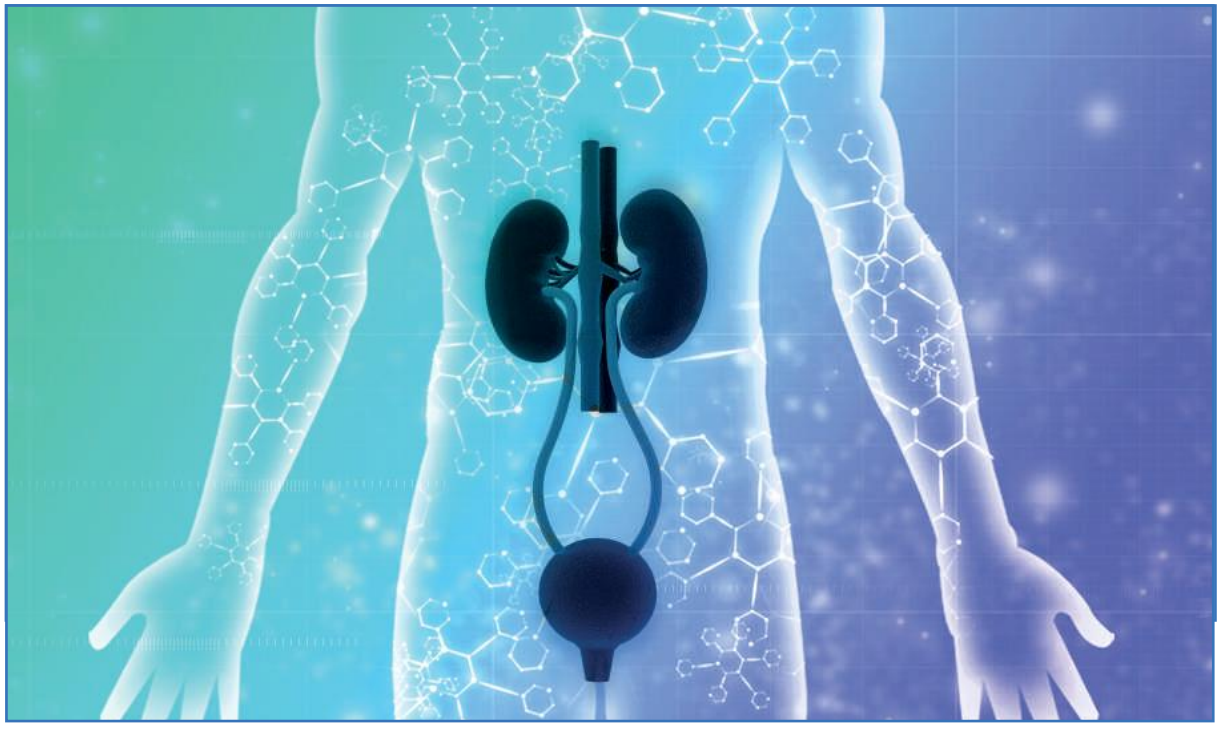
Explain why reduced airflow to the lungs has an impact on metabolism of cells. (5 marks)

Question 10

State how emphysema, pneumonia, vaping and asthma change the rate of gas exchange. (6 marks)

EXCRETORY SYSTEM

14



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The excretory system regulates the chemical composition of body fluids by removing metabolic wastes and regulating water, salts, and nutrients (regulatory processes not required).				
• Deamination of amino acids in the liver produces urea, which then is transported to the kidneys for removal.				
• The nephrons in the kidney facilitate three basic processes: filtration, reabsorption and secretion during urine formation to maintain the composition of body fluids (hormone control is not required).				

The excretory system regulates the chemical composition of the body fluids by removing metabolic wastes and regulating water, salts and nutrients.

Also known as the urinary system or renal system.

(Renal – relating to the kidney, from Latin)

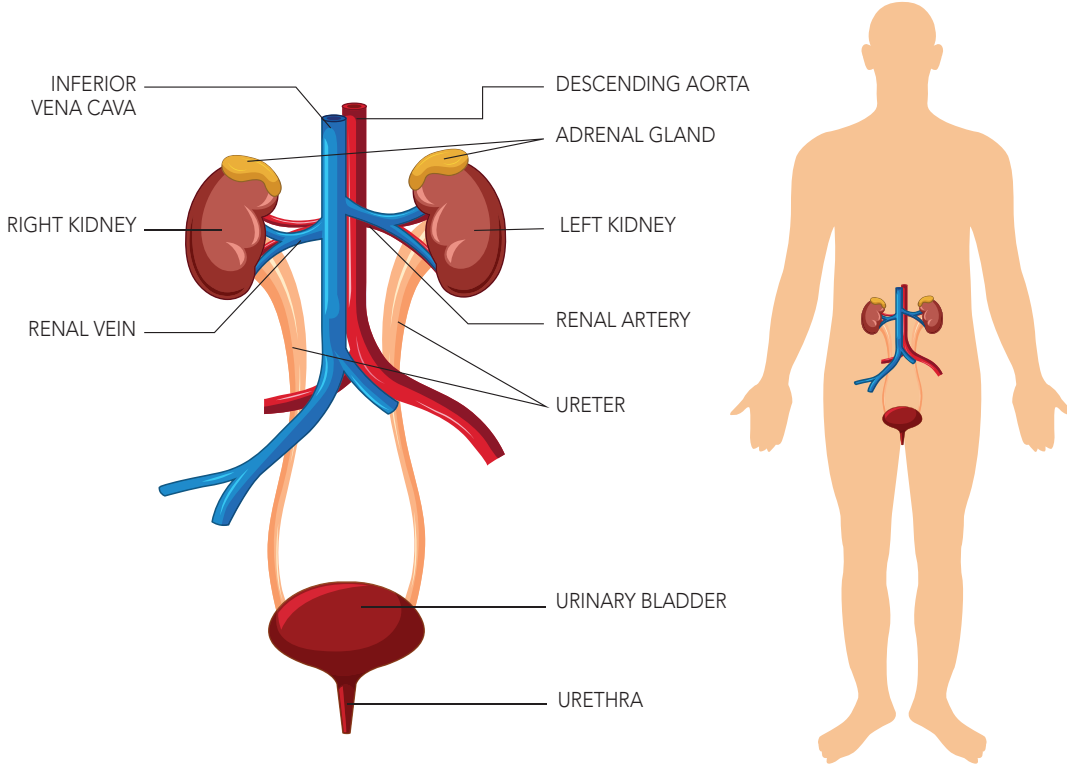


Figure 14.1: The excretory or urinary system

Blood is filtered through the kidney where metabolic wastes and excess materials are removed.

The basic functional unit of the kidney is the **nephrons**.

The nephrons are highly organised within the kidney structure:

- **Renal cortex** – location of the glomerulus, proximal and distal tubule
- **Renal medulla** – location of the loops of Henle and collecting ducts

Urine in the collecting ducts flows into the renal calyses, into the renal pelvis and on into the ureter which leads to the bladder.

The kidneys filter the blood to regulate and optimise the amount, composition, pH and osmotic pressure of body fluids.

Humans are born with an overabundant – or overengineered – kidney capacity. A single kidney with only 75 percent of its functional capacity can sustain life very well.

There are about 1 300 000 nephrons in each human kidney.

Each day, about 180 L of fluid enter into the nephrons of the kidney to be filtered, of which 800–1200 mL of urine is produced. The volume of urine produced depends on the fluid intake and how much water is lost from the body by other means such as sweating or faeces.

Task 14.1: Label the renal calyses (singular: calyx), the renal pelvis, ureter, cortex and medulla in the diagram upper left in Figure 14.2.

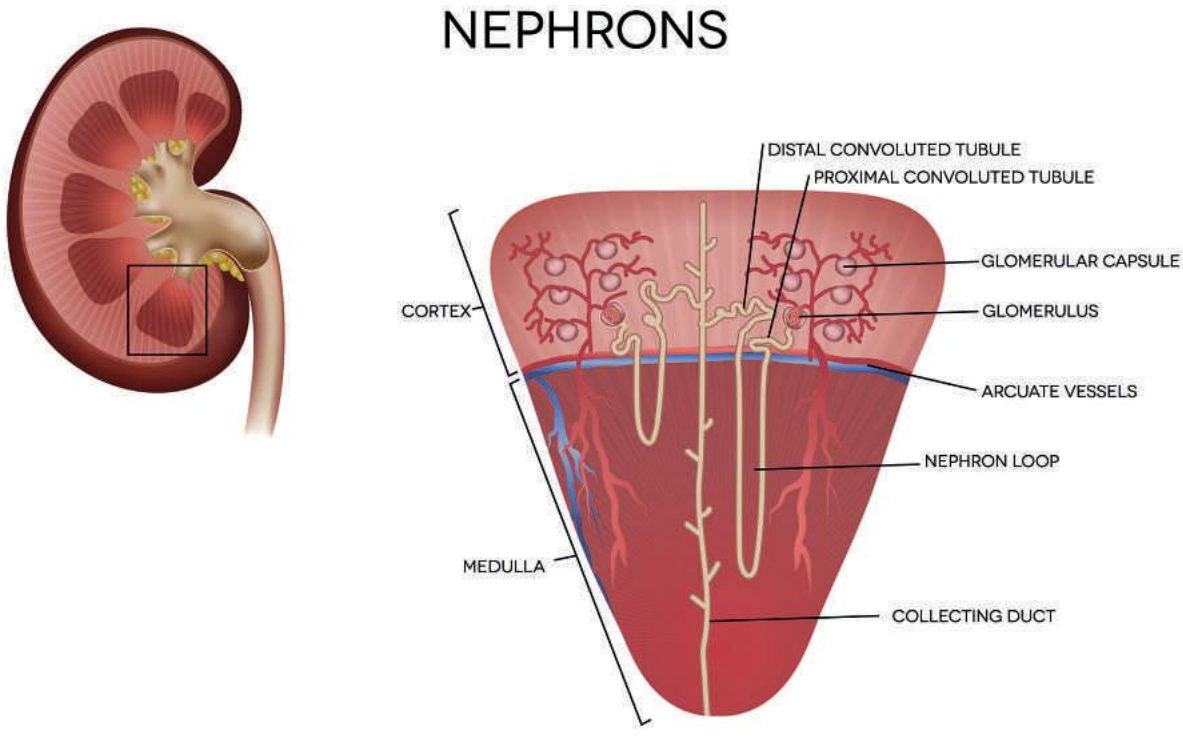


Figure 14.2: Internal structure of the kidney

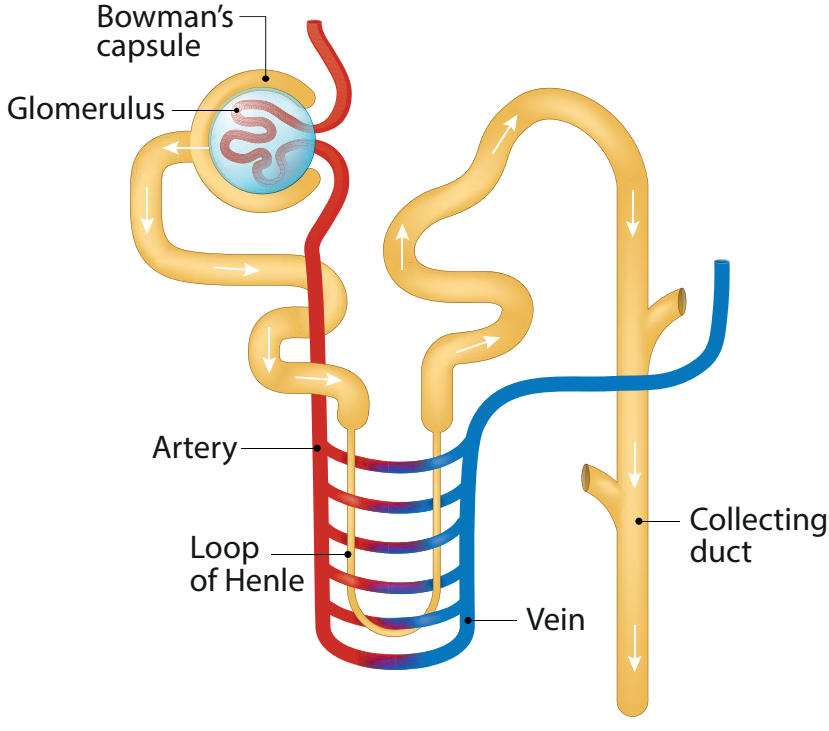


Figure 14.3: The nephron

Task 14.2: Use the names of the parts in Table 14.1 to complete the labelling of Figure 14.3. Complete Table 14.1 by adding the functions of each part listed.

Table 14.1: Parts and function of the nephron and its blood supply

Label	Part	Function
A	Afferent arteriole	
B	Efferent arteriole	
C	Glomerulus	
D	Bowman's capsule/glomerular capsule	
E	Proximal convoluted tubule	
F	Loop of Henle	
G	Distal convoluted tubule	
H	Collecting duct	
I	Renal venule	

Water makes up the largest proportion of the urine.

The body needs to excrete water via the kidneys to remove soluble waste products.

Urine colour is a good indicator of the body's level of hydration.

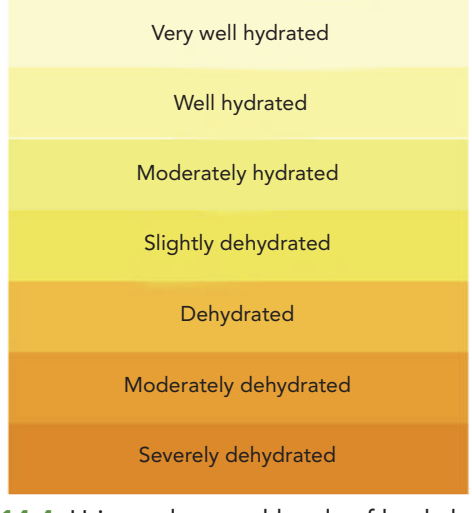


Figure 14.4: Urine colour and levels of body hydration

Drinking too much water causes the body to lose electrolytes (dissolved salts) causing fatigue, restlessness, headaches and feelings of anxiety.

Drinking too little water causes dehydration, which may cause seizures or low blood volume shock or kidney problems.

The nephron is structured to filter the blood to remove unwanted contents and reabsorb useful contents. They are specialised tubules that accomplish the production of urine through the processes of filtration, reabsorption and secretion.

Table 14.2: Processes occurring in the nephron

Process	Description	Location in nephron
Filtration	The process of separating materials in a liquid (blood) through the permeable membrane, based on the size of the particles and under pressure caused by the heart beat	Glomerulus Bowman's capsule
Reabsorption (selective)	Absorption by the nephron of substances that were filtered into the tubules, such as glucose, amino acids, or sodium	Proximal convoluted tubule Loop of Henle
Reabsorption (facultative)	Absorption of materials by hormonal control – water and salts can be removed from the filtrate under the influence of hormones	Distal convoluted tubule Loop of Henle
Secretion	Process adds materials to the filtrate from the blood such as ammonium ions, creatinine and drugs e.g. penicillin; can be active or passive depending on the concentration gradients	Proximal convoluted tubule Distal convoluted tubule Collecting duct

Table 14.3: Comparison of contents of blood plasma and urine

Contents	Plasma	Urine
Water	Constant concentration	Variable amounts
Glucose	Present	Absent
Amino acids	Present	Absent
Urea	Very low concentration	High concentration
Metabolites	Low concentration	High concentration

Most nitrogenous wastes are produced in the liver through the breakdown of excess amino acids – deamination. There are three main forms of nitrogenous wastes produced:

- **Urea** – highly soluble in water and practically non-toxic
- **Ammonia** – highly toxic, especially to the brain, and it requires large amounts of water to remove it from the body; found in vey low concentrations in the urine
- **Creatinine** – is a byproduct of normal muscle function. It is a metabolite of creatine phosphate, which the muscles use as an energy source.

Ammonia is the product of the deamination of proteins in the liver. The nitrogen-containing group NH_4^+ from proteins is changed to NH_3 (ammonia) which then is changed to urea. The body can tolerate higher concentrations of urea than ammonia in the body fluids and it takes much less water to remove urea than ammonia from the body fluids.

Question 1

Indicate the location where in the nephron each of the following occurs.

(a) Filtration _____
(1 mark)

(b) Active reabsorption _____
(1 mark)

(c) Active secretion _____
(1 mark)

(d) Concentration of urine _____
(1 mark)

Question 2

Tests can be done for the presence of glucose and amino acids in urine. What does the presence of these chemicals in the urine indicate? (4 marks)

_____

_____

_____

Question 3

Your kidneys produce urine continuously and your bladder stores it. Without a bladder you would continuously release urine as you produce it, which would certainly be a social problem! Why would health be at risk if humans were to release urine continuously? (2 marks)

_____

_____

Question 4

When your bladder is full, you feel the urge to urinate. Sometimes this is not socially convenient and you put off going to the toilet. Why is it not a good idea to delay urination for too long? (3 marks)

_____

_____

Question 5

A particular health risk exists in females because they have short urethras and they should be especially careful about their personal hygiene habits. Explain this risk and the precautions that could be taken. (3 marks)

Question 6

Explain the effect of each of the following on the volume and concentration of urine.

(a) Drinking a large amount of water (1 mark)

(b) Eating a very salty meal (1 mark)

(c) A hot, dry day (1 mark)

(d) A cold winter's day (1 mark)

Question 7

On average about 180 L of plasma filtered through a kidney in 24 hours during normal conditions. The kidney receives about 180 L of blood (about 90 L of plasma) per day. A person's body contains about 100 mL of blood per kilogram of body weight.

Calculate how many times your blood is filtered per day. (3 marks)

15 SKELETON

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The skeletal framework of the body consists of bone and cartilage which function to provide body support, protection and movement, and is facilitated by the structure and function at cell and tissue levels.				

SKELETON

The skeleton:

- is the internal framework of the body composed of 206 bones in adults. Babies have more bones, but some fuse together during infancy and childhood
- determines the overall shape of the body and provides anchorage for muscles and internal organs
- has many functions that overlap with the functions of other systems.

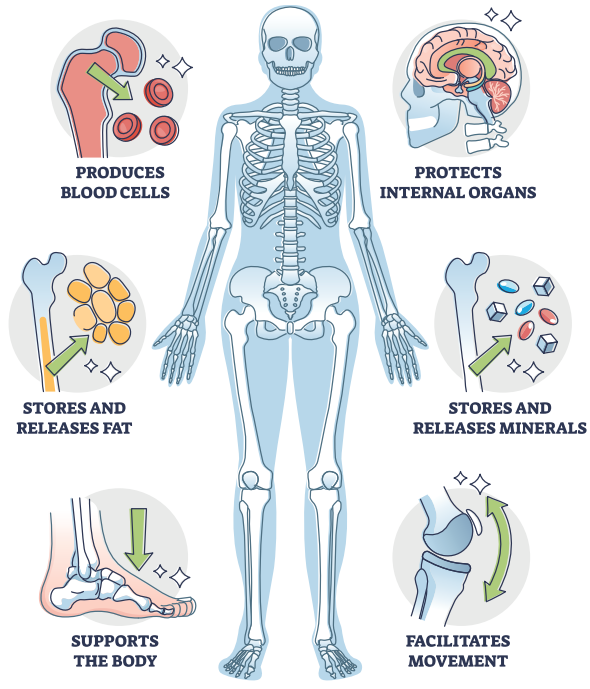


Figure 15.1: Functions of the skeletal system

The skeleton can be classified into two sections.

- **Axial skeleton** – consists of the vertebral column, the rib cage, the skull and other associated bones forming the axis of the body. It is mainly involved in protection of internal organs.
- **Appendicular skeleton** – consists of the shoulder (pectoral) girdle, the pelvic girdle, and the bones of the upper and lower limbs i.e. the appendages (arms and legs) and the girdles attaching them to the axial skeleton attachments. It is mainly involved in movement of the body.

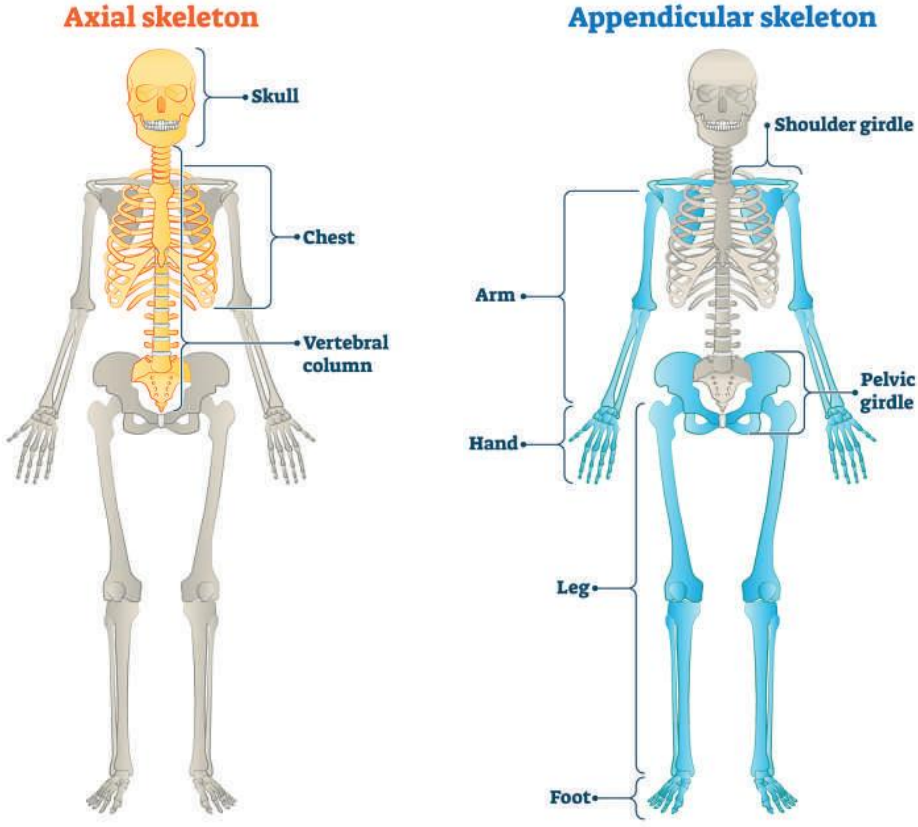


Figure 15.2: The axial and appendicular skeleton sections

Task 15.1: Using Figure 15.2 draw a table to list the bones in the axial and appendicular skeletons.

DIFFERENTIATING BETWEEN MALE AND FEMALE SKELETONS

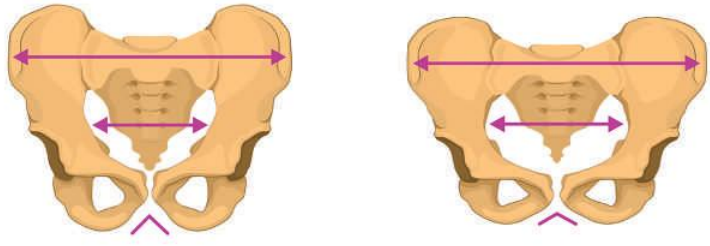


Figure 15.3: Male and female pelvic girdles

Differences between the male and female pelvis:

- males have a generally narrower pelvis than females
- the pelvic opening in males is smaller than that of females
- the angle formed between the pubic bones (pubic arch) is greater than 90° in females and less than 90° in males
- females usually have lighter and thinner bones in the pelvis

The reason for the difference in the structure of the pelvis in females is for the purpose of childbearing.

STRUCTURE OF BONES

Bone tissue is an example of **connective tissue**. The distinguishing feature of bone as a connective tissue is its hard matrix between the cells (osteocytes).

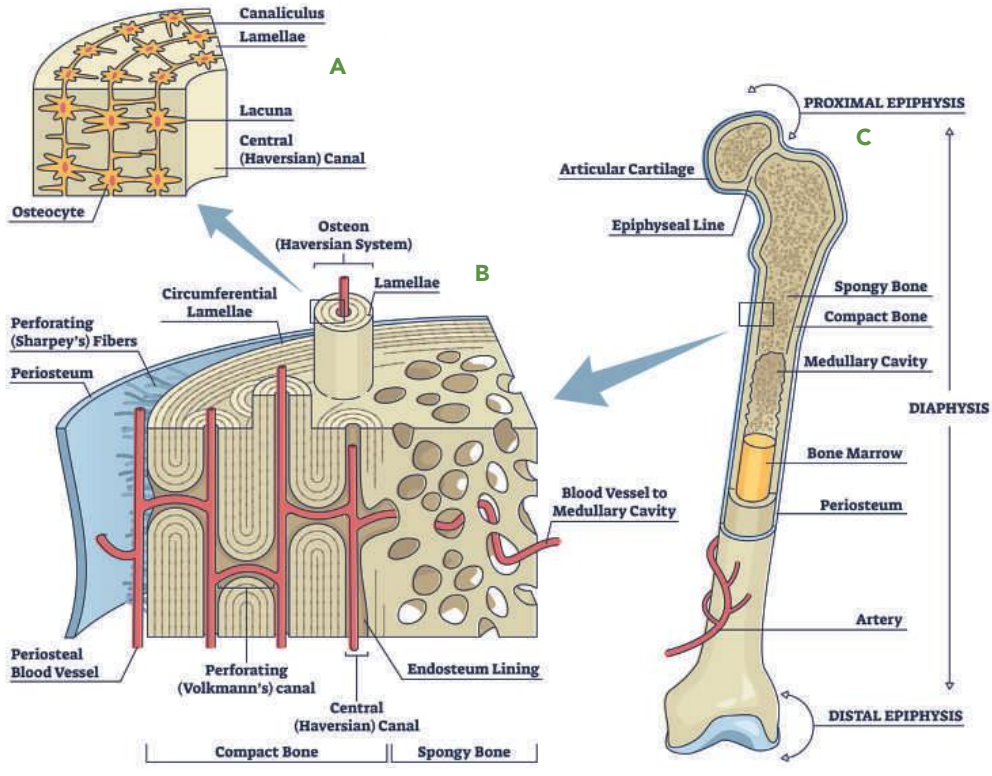


Figure 15.4: Structure of a long bone and bone tissue

BONE TISSUE

- osteocytes – living bone cells
- lacunae – fluid filled spaces in which the osteocytes sit
- canaliculus – fluid-filled connections between osteocytes and the central Haversian canal
- lamellae – layers of bone matrix produced by the osteocytes
- Haversian canal – fluid-filled space holding the blood vessels and nerves servicing the bone tissue

Nutrients and wastes move to and from the blood vessels in the Haversian canal to the osteocytes via the canaliculi by diffusion. This is a slow process, so the metabolic rate of bone tissue is low, hence the slow rate of repair of broken bones.

TYPES OF BONE TISSUE

- **Compact bone** – contains dense lamellae in a circular pattern around a central Haversian canal called an osteon; the only spaces present are for the blood vessels and nerves. This type of bone provides strength and protection against knocks.
- **Spongy bone** – is composed of lattice-like arrangement of osteocytes known as trabeculae. Trabeculae form a mesh-like network of bony tissue that are aligned along regions of biomechanical stress. The overall structure looks like a sponge where the holes are surrounded by living tissue. The holes are filled with adipose tissue or red bone marrow.

OVERALL STRUCTURE OF A TYPICAL LONG BONE

- epiphyses – proximal – nearer to the body axis; distal – out end of the bone
  - location of joints with other bones
  - made up of spongy bone
  - contains red bone marrow
- epiphyseal line – between the epiphysis and the diaphysis
  - made of cartilage
  - permits growth and lengthening of the bone, as the cartilage reproduces and ossifies
- diaphysis – is the shaft of the long bone
  - made up of compact bone surrounding a space called the medullary cavity
  - medullary cavity contains yellow bone marrow
  - is very strong
  - provides sites for the attachment of muscles
- periosteum – membrane surrounding the long bone
- endosteum – membrane surrounding the inside of the medullary cavity

ARTICULAR CARTILAGE

Articular cartilage:

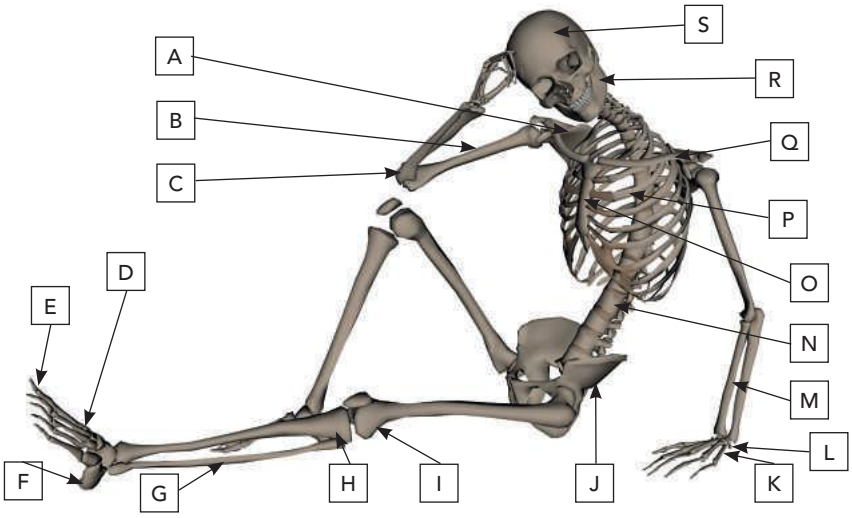
- is made of hyaline cartilage
- is smooth and shiny
- is found on the ends of long bones where they articulate with other bones in joints
- lacks direct blood supply and nerves – materials are exchanged directly with the surrounding joint fluids, hence to slow rates of repair to these cartilages if damaged
- reduces friction in joint movement
- absorbs biomechanical forces (shock absorber)
- stabilises the joint.

Bone words based on *osteo* meaning bone in Latin:

- ossification – the process of bone formation
- osteocyte – bone cell
- periosteum – membrane surrounding bone
- osteitis – inflammation of bone
- osteomyelitis – bacterial/fungal bone infection

Question 1

Label the bones of the skeleton labelled on the diagram below. (1 mark each)



Part	Name	Part	Name
A		K	
B		L	
C		M	
D		N	
E		O	
F		P	
G		Q	
H		R	
I		S	
J			

Question 2

Could you live without either your axial or appendicular skeleton? Justify your answer. (3 marks)

Question 3

You have been given a microscopic slide of bone tissue. Describe how you would determine if it was spongy bone or compact bone. (2 marks)

Question 4

Describe how the shoulder girdle and the pelvic girdle are joined to the axial skeleton. (4 marks)

Question 5

Draw up a table to compare the structure of the arm and the leg. (13 marks)

Question 6

Compare the structure of the hand and the foot and explain the differences. (5 marks)

Question 7

State the function of the clavicle. (3 marks)

Question 8

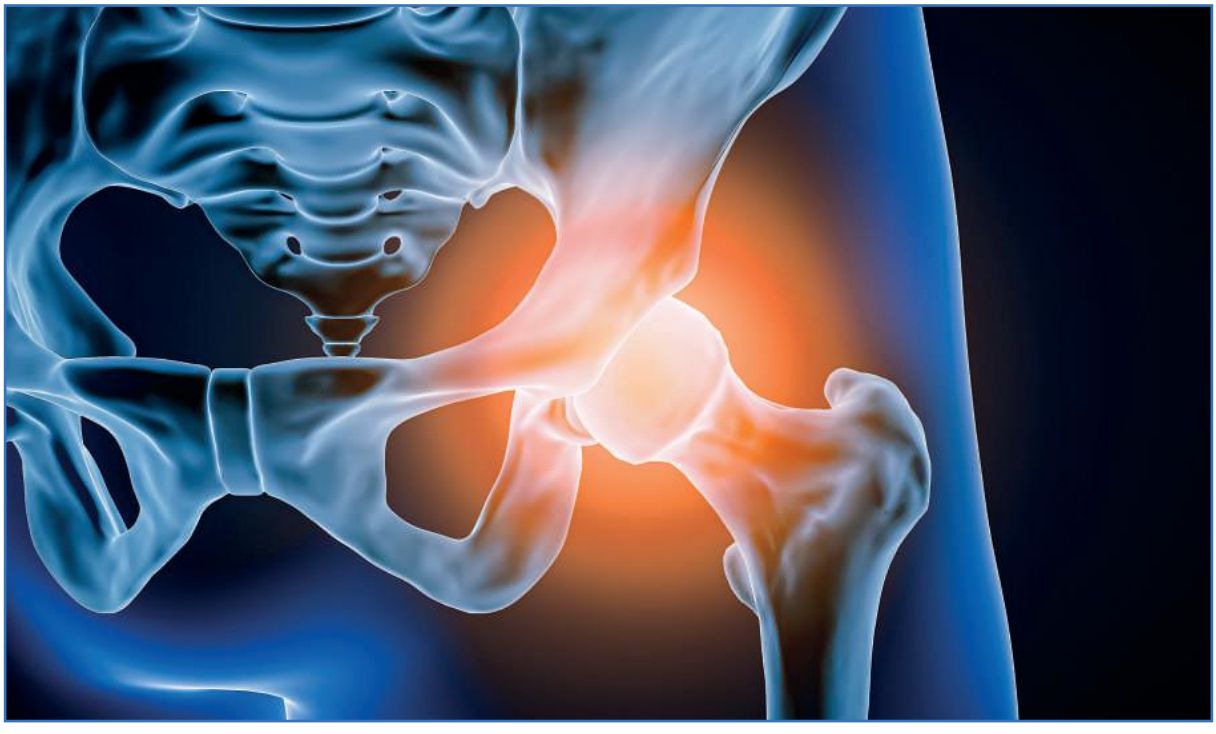
What are the ossicles and describe where they can be found. (1 mark)

Question 9

Explain why the ends of the long bones are covered with cartilage. (2 marks)

JOINTS

16



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Articulations of joints of the skeleton are classified according to their structure or the range of movements permitted.				
• Osteoporosis and osteoarthritis are diseases, primarily of ageing, that cause disability. Increased understanding of the causes of these conditions leads to improved practices for management and prevention.				
• Treatment of conditions due to system or organ dysfunction has changed through improvements in early diagnosis and appropriate use of drugs, physical therapy, and removal and/or replacement of affected parts.				

TYPES OF JOINTS

Without joints, the body could not make movements such as walking or waving the arms, shaking the head or chewing.

Joints are where bones articulate with one another. There are three main types of joints.

- **Immoveable joints** (fibrous joints) – bones are held closely together by fibrous material with no joint cavity allowing very little if any movements between the bones. These include:
  - sutures between the bones of the skull
  - syndesmosis between the radius and ulna in the forearm and the tibia and fibula in the ankle area.
  - gomphosis between the teeth and the jaw bones.
- **Slightly moveable joints** (cartilaginous joints) – where the movement of the bones in the joint is limited by fibrous tissue or cartilage and there is no joint cavity. These include:
  - intervertebral discs between the vertebrae
  - pubis symphysis between the two pubic bones of the pubic arch.
- **Freely moveable joints** (synovial joints) – have a fluid-filled joint cavity between the articulating bones.

SYNOVIAL JOINTS

Types of synovial joints:

- **Pivot joint**: One bone is encircled by a ring formed by the other bone at the joint and a ligament. The bone that pivots may either rotate within the ring or the ring may rotate around the bone. e.g. between the first and second cervical vertebrae. This joint permits rotational movement around a single axis.
- **Hinge joint**: Movement is limited to bending and straightening movements along one plane e.g. elbow, knee, ankle, and joints between the phalanges.
- **Condyloid or ellipsoid joint:** One of the bones has an oval-shaped, or convex, end that fits into the depressed oval-shaped, or concave end of another bone e.g. found between the radius and bones of the wrist. The movements allowed by this type of joint include bending and straightening, side-to-side, and circular movements.
- **Saddle joint:** The bones at these joints form what looks like a rider on a saddle. One bone is turned inward at one end, while the other is turned outward e.g. the thumb joint between the thumb and palm. These joints are very flexible, allowing for bending and straightening, side-to-side, and circular movements.
- **Plane joint:** Bones are of similar size and the surfaces where the bones meet at the joint are nearly flat e.g. found between bones of the wrist and foot and between the clavicle and scapula. Bones in plane joints slide past each other in a gliding motion.
- **Ball-and-socket joint:** The end of one bone at this type of joint is rounded (ball) and fits into the cupped end (socket) of another bone e.g. hip and shoulder joints. These joints allow the greatest degree of motion allowing bending and straightening, side-to-side, circular, and rotational movement.

Each of the different types of synovial joints allows for specialised movements that permit different degrees of motion.

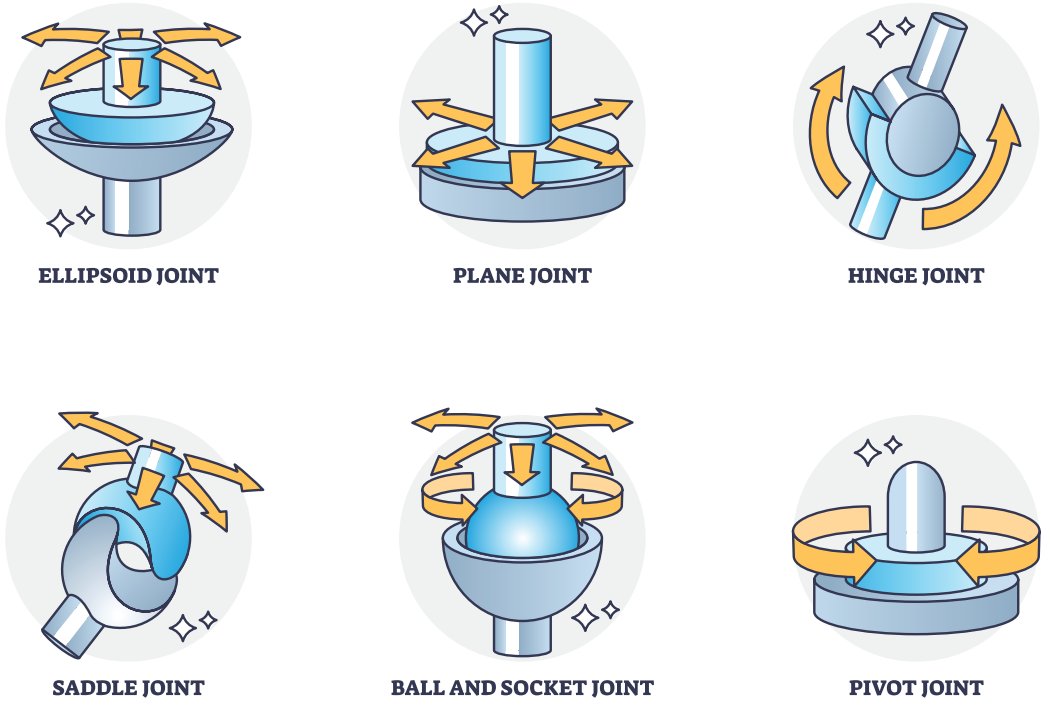


Figure 16.1: Movement at synovial joints

TYPES OF MOVEMENTS AT JOINTS

Table 16.1: Descriptions of the types of movements at joints

Type movement	Description
Flexion	Decreasing the angle between two bones of the joint; bending the limb
Extension	Increasing the angle between two bones in the joint; straightening the limb
Pronation	Moving the body part so the ventral or front surface faces downward; palm facing the ground
Supination	Moving the body part so the ventral or front surface faces upwards; palm facing upwards
Abduction	Moving the limb away from the midline of the body of the body
Adduction	Moving the limb towards the midline of the body
Circumduction	Moving the limb so the outer end moves in a circle
Internal rotation	Turning the limb in towards the midline of the body
External rotation	Turning the limb outwards away from the midline of the body
Dorsiflexion	Flexion of the foot and toes upwards
Plantar flexion	Flexion of the toes and foot downwards

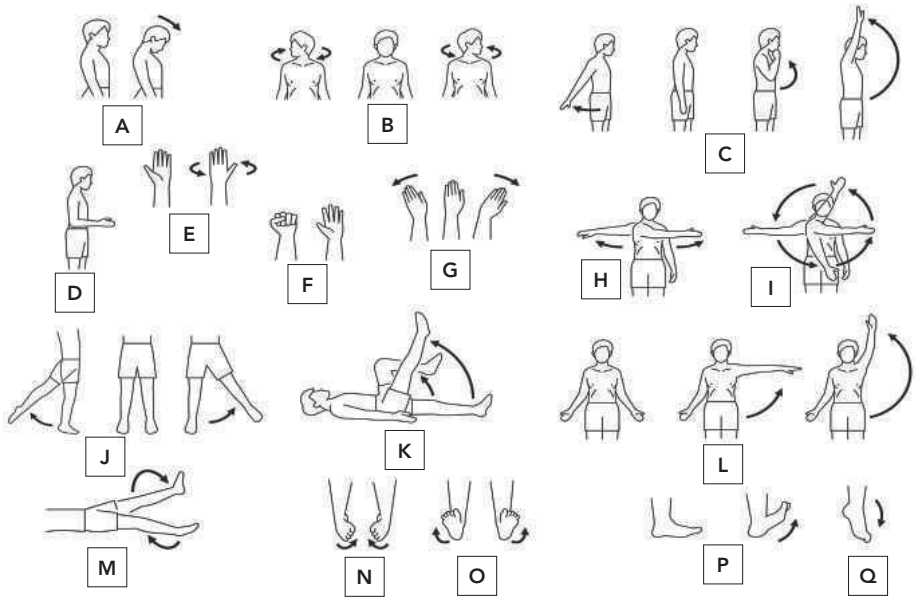


Figure 16.2: Movement at joints

Task 16.1: Use the information in Table 16.1 to determine the type of movement occurring in the illustrations in Figure 16.2.

SYNOVIAL JOINT STRUCTURE

Features of the synovial joint that **reduce friction** between articulating bones are:

- articular cartilage
- synovial fluid.

Features of the synovial joint **increase stability** of the joint are:

- tendons and ligaments surrounding the joint
- joint capsule.

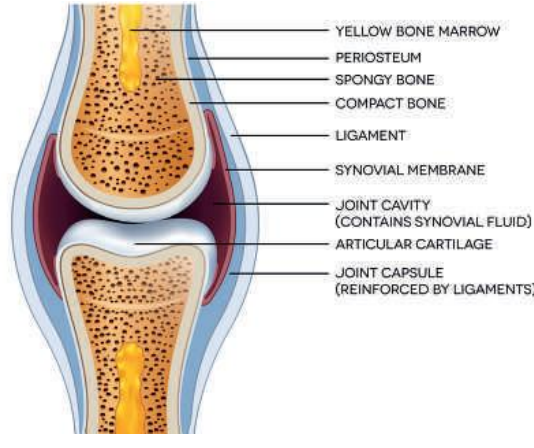


Figure 16.3: Structure of a synovial joint

Bone never touches bone in a properly functioning synovial joint. If it does, then serious damage has occurred and the joint will not function as is should and will cause considerable pain at the site.

DYSFUNCTION OF THE SKELETON

STRAIN VS SPRAIN

- Strain is injury to the tendons and muscles controlling the movement of the joint.
- Sprain is damage to the ligaments that join bones to bones.
- Both caused by over extension of the movements of the joint causing damage to surrounding tissue.
- Both cause swelling of the joint.

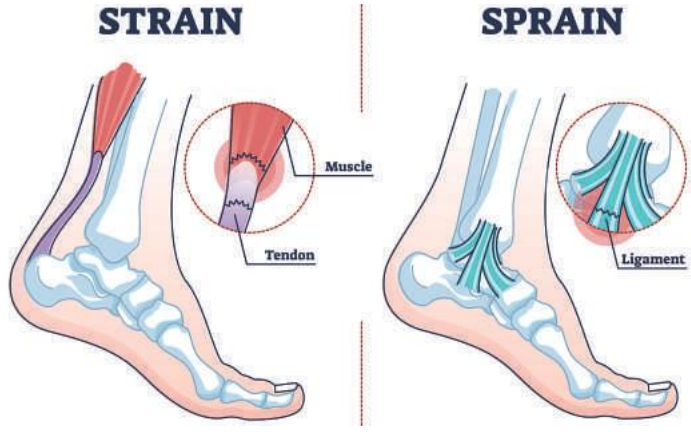
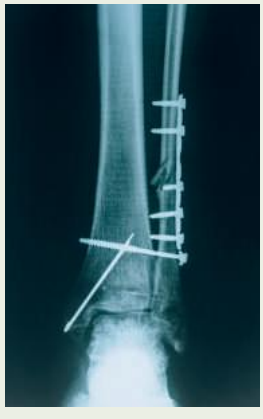


Figure 16.4: Strain (left) vs sprain (right)

BROKEN BONES

Task 16.2: Circle the point of the break in this X-ray.



Bone breaks or fractures need to be stabilised to allow the area of the break to grow new bone tissue.

Depending on the location and severity of the break, a broken limb needs to be in a cast from 3 weeks to 3 months.

External and internal splints may be used to support the broken pieces to help the healing process.

Figure 16.5: X-ray of plates and screws used to fix a broken bone

OSTEOPOROSIS AND OSTEOARTHRITIS

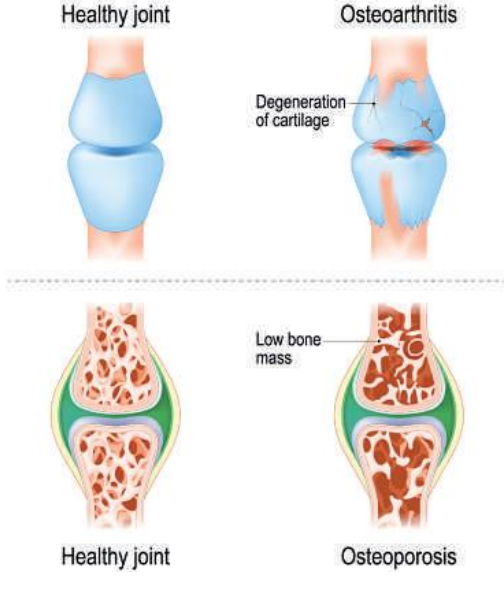


Figure 16.6: Comparing healthy, osteoporotic and osteoarthritic bones

Osteoporosis – breakdown of the bone structure making it more likely to break.

Risk factors:

- Gender – women are much more likely to develop osteoporosis than are men.
- Age – the risk of osteoporosis increases with age.
- Ethnic background – people of Caucasian or Asian descent have the greatest risk for osteoporosis.
- Family history – having a parent or sibling with osteoporosis puts a person at greater risk, especially if a parent fractured a hip.
- Body frame size – men and women who have small body frames tend to have a higher risk because they may have less bone mass to draw from as they age.

Hormone levels

Osteoporosis is more common in people who have too much or too little of certain hormones in their bodies. Examples include lower oestrogen levels in post-menopausal women or an overactive thyroid.

Dietary factors

Osteoporosis is more likely to occur in people who have:

- **low calcium intake** affecting bone density
- **eating disorders** which severely restrict food intake

Long-term use of oral or injected corticosteroid medications (steroids) can also increase the risk of osteoporosis.

Lifestyle choices

Some habits can increase your risk of osteoporosis. Examples include:

- **sedentary lifestyle**
- **excessive alcohol consumption**
- **tobacco use.**

Osteoarthritis – breakdown of the cartilage in joints making movement at the joints painful and limited.

Risk factors

- **Older age** – the risk of osteoarthritis increases with age.
- **Gender** – women are more likely to develop osteoarthritis, though it isn't clear why.
- **Obesity** – increased weight adds stress to weight-bearing joints, such as your hips and knees. Also, fat tissue produces proteins that can cause harmful inflammation in and around joints.
- **Joint injuries** – workplace, sporting, or other accidental injuries can increase the risk of osteoarthritis.
- **Repeated stress on the joint** – work or sport places repetitive stress on a joint, which may eventually develop osteoarthritis.
- **Heredity** – some people inherit a tendency to develop osteoarthritis.
- **Metabolic diseases** – including diabetes.

Question 1

Explain the limitations of in the movement at joints. (2 marks)

Question 2

How does your lifestyle in adolescence influence your chances of having osteoporosis later in life? (5 marks)

Question 3

Complete the following table as a summary of comparison of the joint types. (15 marks)

Location	Joint type	Bones in joint	Range of movements
Elbow			
Ankle			
Wrist			
Base of thumb			
Neck			

Question 4

The hip and the shoulder are both ball and socket joints. Contrast these two joints with respect to the bones forming the joint, the strength of the joint and the range of movements capable of each joint. (15 marks)

Question 5

Outline the types of joints found in the skull and describe how each is suited to their location and purpose. (12 marks)

Question 6

The knee has structures called bursae. They are filled with fluid. What is their function? (2 marks)

MUSCLES

17

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The muscular system is organised to maintain posture and produce movement; muscle fibre contraction can be explained using the sliding filament theory.				
• Movement results from the actions of paired muscles, with others acting as stabilisers, to produce the required movement.				

Skeletal muscles work with the skeleton to bring about movement and maintain posture.

Muscle tissue is attached to the skeleton by tendons.

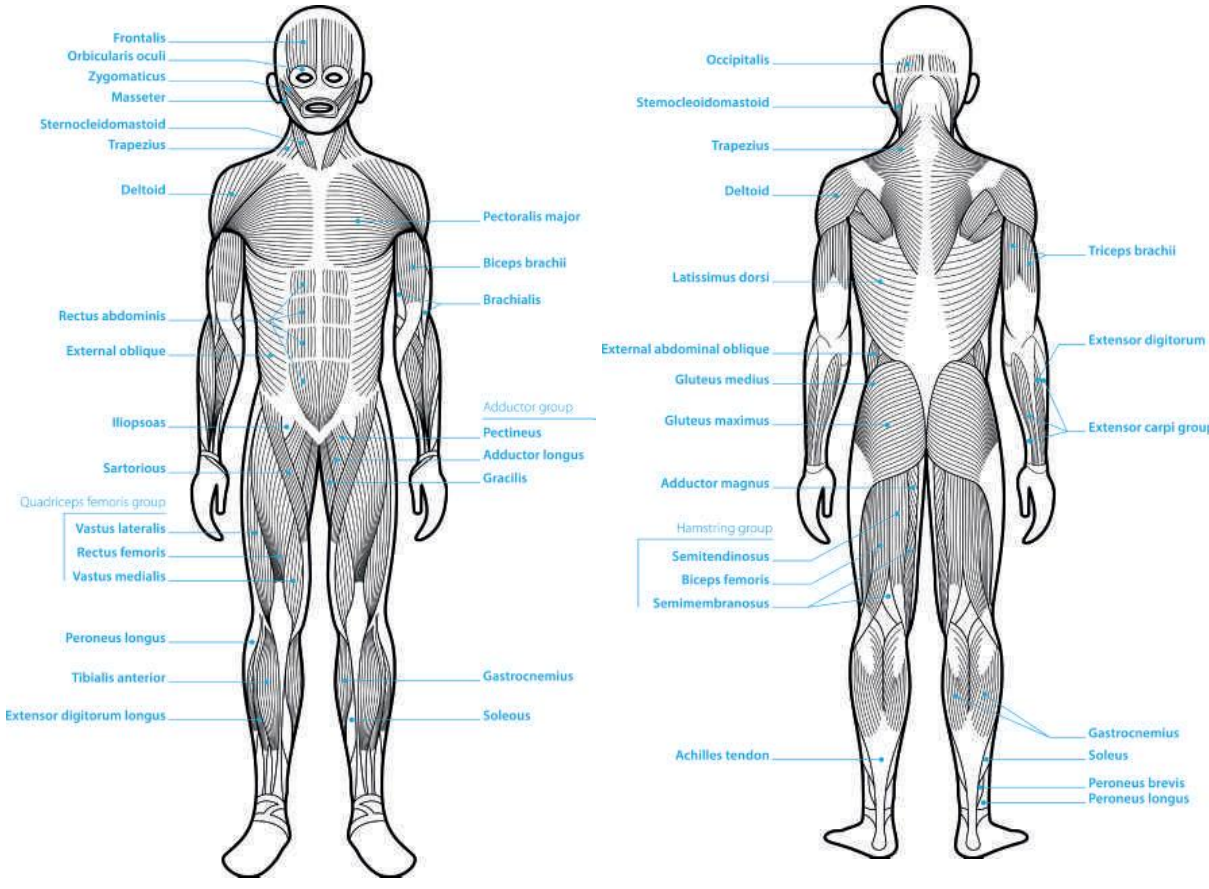


Figure 17.1: Major muscles of the body

Muscles are arranged in opposing (antagonistic) pairs at joints to bring about opposing actions e.g. flexors and extensors; abductors and adductors because **muscles can only contract to produce a pulling force on bones**. Muscles can't push.

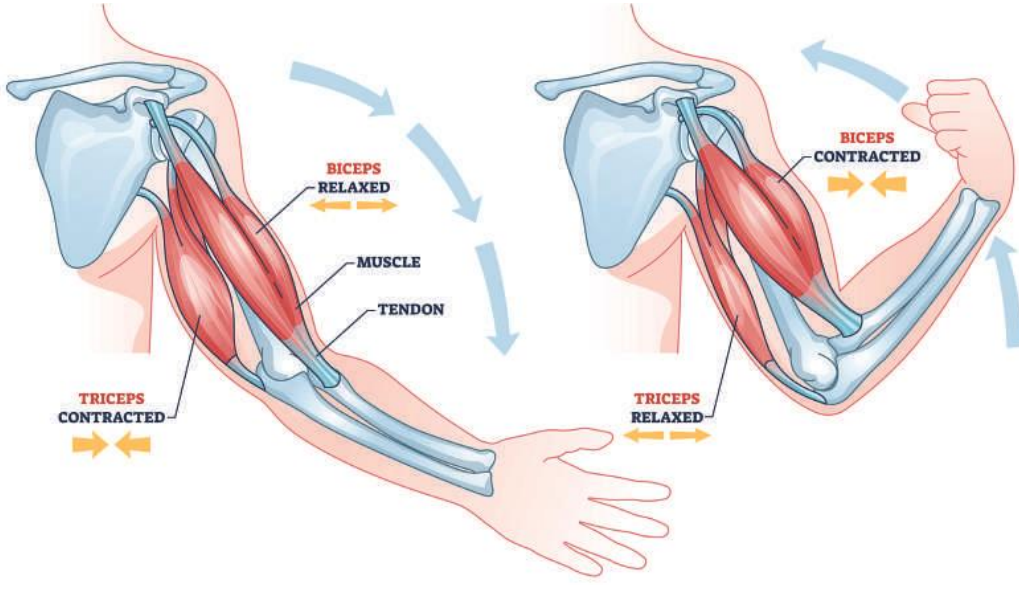


Figure 17.2: Extension and flexion of the elbow caused by antagonistic muscles: the biceps and triceps

The muscle causing the required action is called the **agonist**.

The muscle having the opposite action is called the **antagonist**.

Muscles which steady the joint to prevent unwanted movements are called **synergists**.

Skeletal muscle is organised at different levels to bring about co-ordinated action of the whole muscle.

Muscle is made up of individual **fascia** surrounding a bundle of **muscle fibres** which consist of many **myofibrils** that contain strands of **actin and myosin** proteins in specific arrangements.

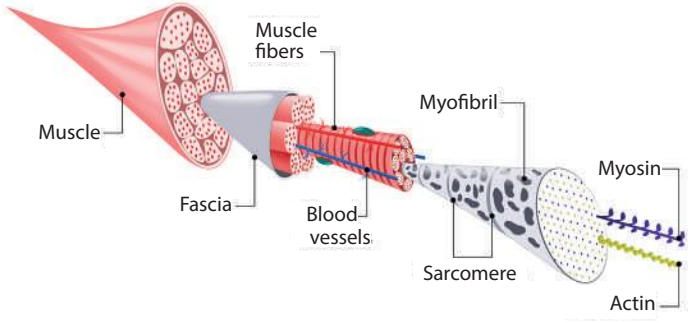


Figure 17.3: Levels of organisation of muscle

Movement at the microscopic level brings about movement at the body level by all sarcomeres in the myofibrils working together.

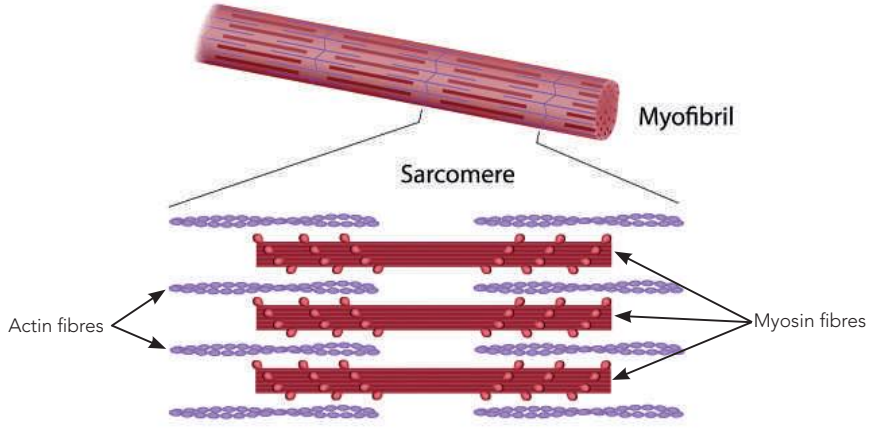


Figure 17.4: Arrangement of actin and myosin in the sarcomere of a myofibril

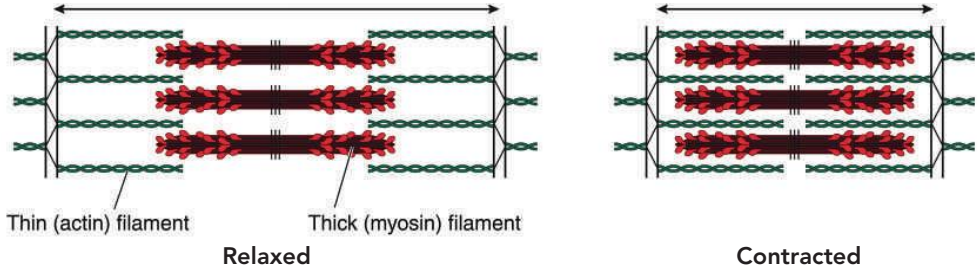


Figure 17.5: Arrangement of actin and myosin filaments when the muscle is relaxed and contracted

The shortening of the sarcomere causes shortening of the muscle fibre and ultimately the whole muscle. With the muscle attached to the bones, this shortening causes movement at the joint.

The strength and power of a muscle depends on how many myofibrils it has. Muscles come in a great variety of sizes. The largest muscle is in the buttocks whereas the smallest is in the ear.

Muscle tone: sustained, small contractions give a firmness to a relaxed muscle without producing movement. This is essential for maintain posture e.g. the head is kept in an upright position due to the small contractions of the neck muscles to balance the skull on the vertebral column.

Question 1

For each of the muscle actions listed, state the opposite action. (4 marks)

(a) Flexion _____

(b) Adduction _____

(c) Pronation _____

(d) Dorsiflexion _____

Question 2

Describe the structure of tendons and why they are suitable for attaching muscle to bone. (2 marks)

_____

_____

_____

Question 3

State the muscles that bring about the following actions. (6 marks)

(a) Pointing the toes _____

(b) Bending the knee _____

(c) Lifting the shoulders (as in a shrug) _____

(d) Abducting the humerus _____

(e) Extending the leg backwards _____

(f) Straightening the elbow _____

Question 4

Explain the location of the muscles controlling the movement of the fingers. (2 marks)

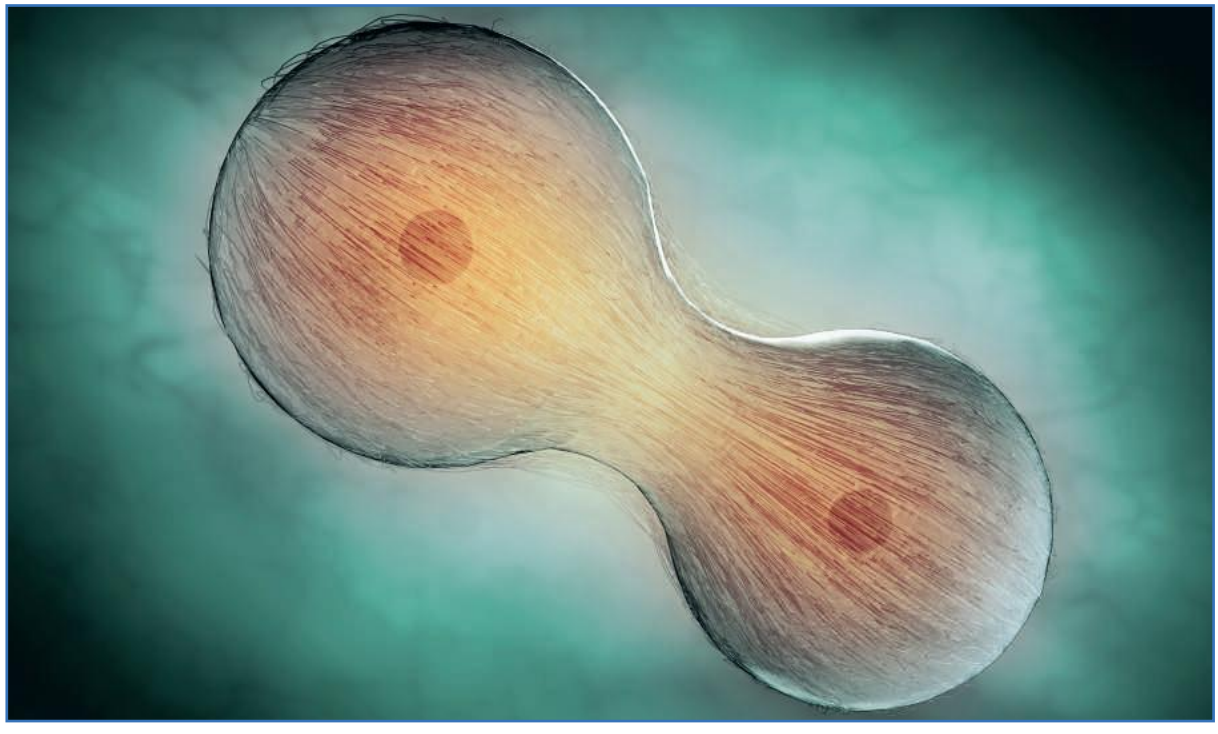
Question 5

Rigor mortis is the stiffening of joints and muscles after death. Describe the state of the muscle myofibrils in this condition. (2 marks)

Question 6

Explain why each skeletal muscle is attached to at least two bones. (2 marks)

18 MITOSIS AND MEIOSIS



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Mitosis forms part of the cell cycle producing new cells with the same genetic content.				
• Meiosis produces gametes for reproduction and involves DNA replication, chromosome pairing, and two successive nuclear divisions distributing haploid sets of chromosomes to each gamete.				
• Differences between mitosis and meiosis reflect their roles in the body.				
• Crossing over, non-disjunction and random assortment of chromosomes during meiosis will produce gametes with different genetic content.				

Cells can undergo two types of cell division: **mitosis** and **meiosis**.

Both types of cell division involve the doubling of the genetic material – DNA – and the separation of chromosomes.

Chromosomes become visible during prophase in mitosis and prophase 1 in meiosis, as the DNA strands and histone proteins condense and coil. At the end of cell division, the strands of DNA uncoil and spread throughout the nucleus of the cell.

Double chromosomes appear in Stage S in the cell cycle, after the duplication of DNA.

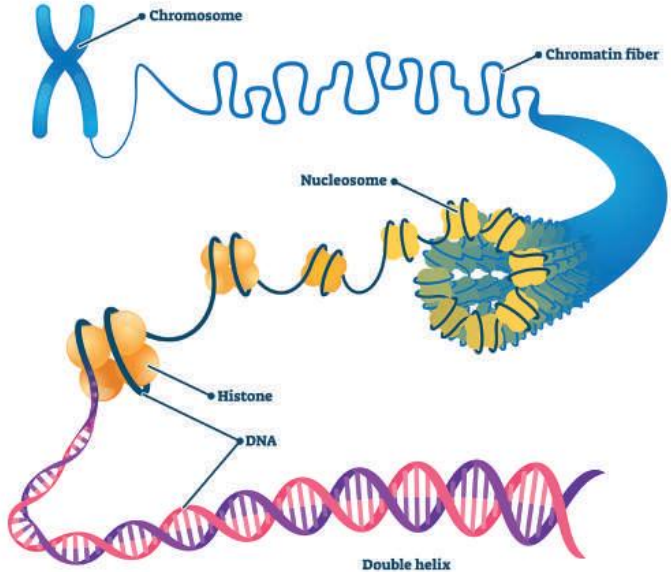


Figure 18.1: Relationship between chromosomes and DNA

Chromosomes can be single stranded as at the end of mitosis or double stranded (as shown in Figure 18.2) at the beginning of mitosis.

Double stranded chromosomes consist of two chromatids. When the chromatids separate, they are called chromosomes.

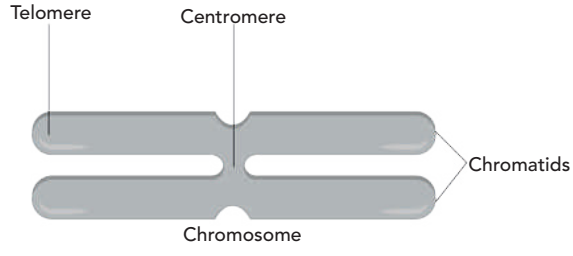


Figure 18.2: Chromosome compared to chromatid

The cell cycle is the series of events that take place in a cell as it grows and divides to produce daughter cells.

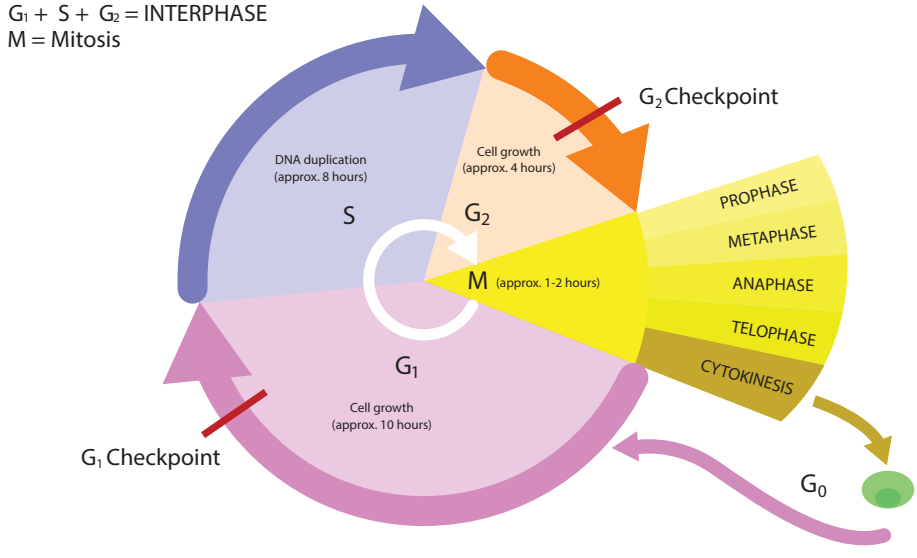


Figure 18.3: Cell cycle

MITOSIS

- cell division that results in two daughter cells, each having the same number and kind of chromosomes as the parent nucleus, typical of ordinary tissue growth.

ROLES OF MITOSIS

- **Growth and development** – to produce more cells from the zygote to increase the size of the body and allow cells to differentiate into all the tissues required in the body.
- **Cell replacement** – to repair tissues that are damaged such as a cut, blister or pimple.

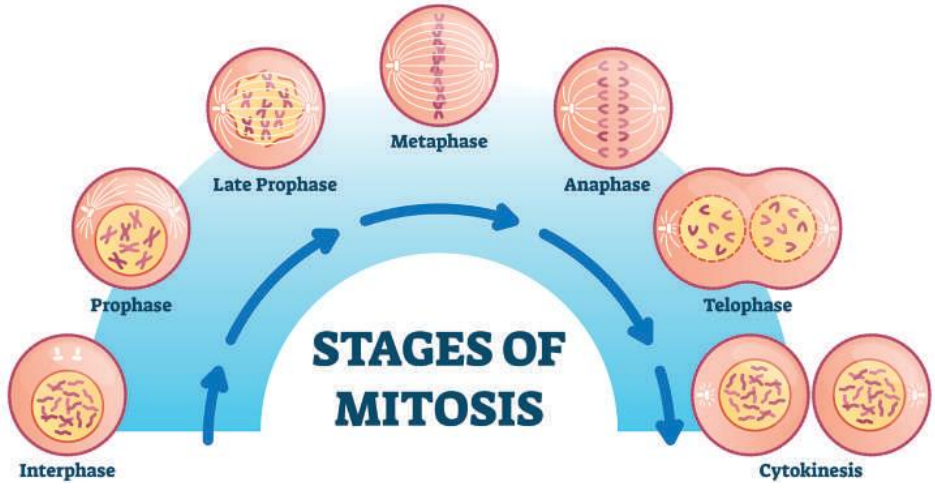


Figure 18.4: Stages of mitosis

Task 18.1: Complete the following table to describe the activities in the cell during each phase of mitosis.

Stage of mitosis	Cellular activities
Interphase	
Prophase	
Metaphase	
Anaphase	
Telophase	
Cytokinesis	

Task 18.2: Identify different stages of mitosis in cells in Figures 18.5 and 18.6.

Figure 18.5: Onion cells

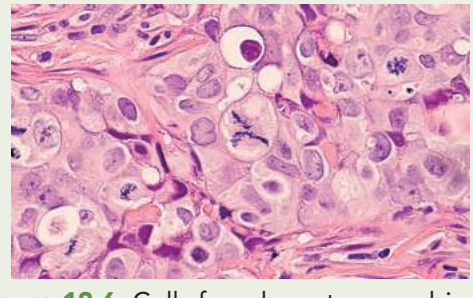


Figure 18.6: Cells from breast cancer biopsy

MEIOSIS

- Meiosis is cell division that results in **four** daughter cells each with **half** the number of chromosomes of the parent cell.
- It involves two divisions of the cell.

ROLE OF MEIOSIS

Meiosis is for the production of gametes for sexual reproduction.

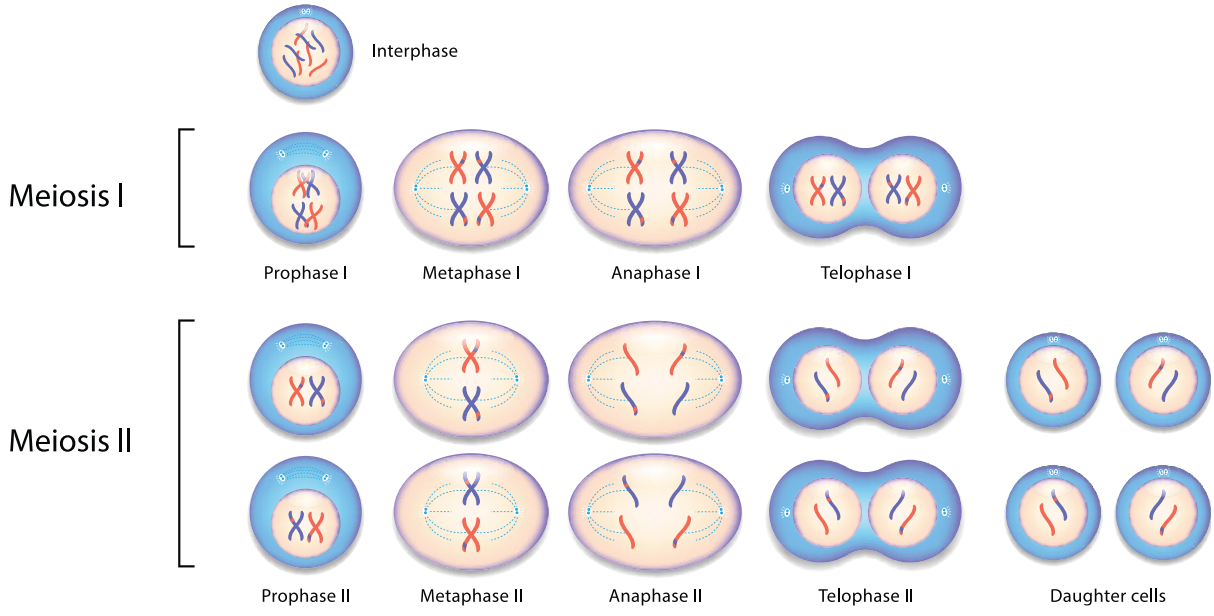


Figure 18.7: Stages of meiosis cell division

Chromosomes occur in pairs in cells – one from the female parent and one from the male parent. These are called **homologous chromosomes**. They carry the same genetic information but may be of different variations, so the cells have two copies of the genetic information (except for the sex chromosomes, X and Y).

Meiosis involves the **pairing of homologous chromosomes** during the first phase of meiosis.

During this pairing, the chromatids of the homologous chromosomes may intertwine – remember, the contents of cells are always in motion. Where the two chromatids come contact, there may be a swapping of sections. Each still ends up with the same amount of DNA, but not on the same chromatid. The crossing over point is called the **chiasma**. This is important in that the resulting chromatids distributed to the daughter cells are all different, producing variations in the offspring arising from these gametes.

Look carefully at the daughter cells of meiosis in Figure 18.7. Crossing over has occurred in Metaphase 1, producing daughter cells with different genetic contents.

The number of chromosomes in a cell is designated using the letter N.

N – one set of chromosomes from one parent or the **haploid** number as found in gametes.

2N – two sets of chromosomes, one from each parent or the **diploid** number, as found in body cells.

The processes of **meiosis** ensures the **gametes have one of each of the pair** of chromosomes found in the parent cell.

COMPARING MITOSIS AND MEIOSIS

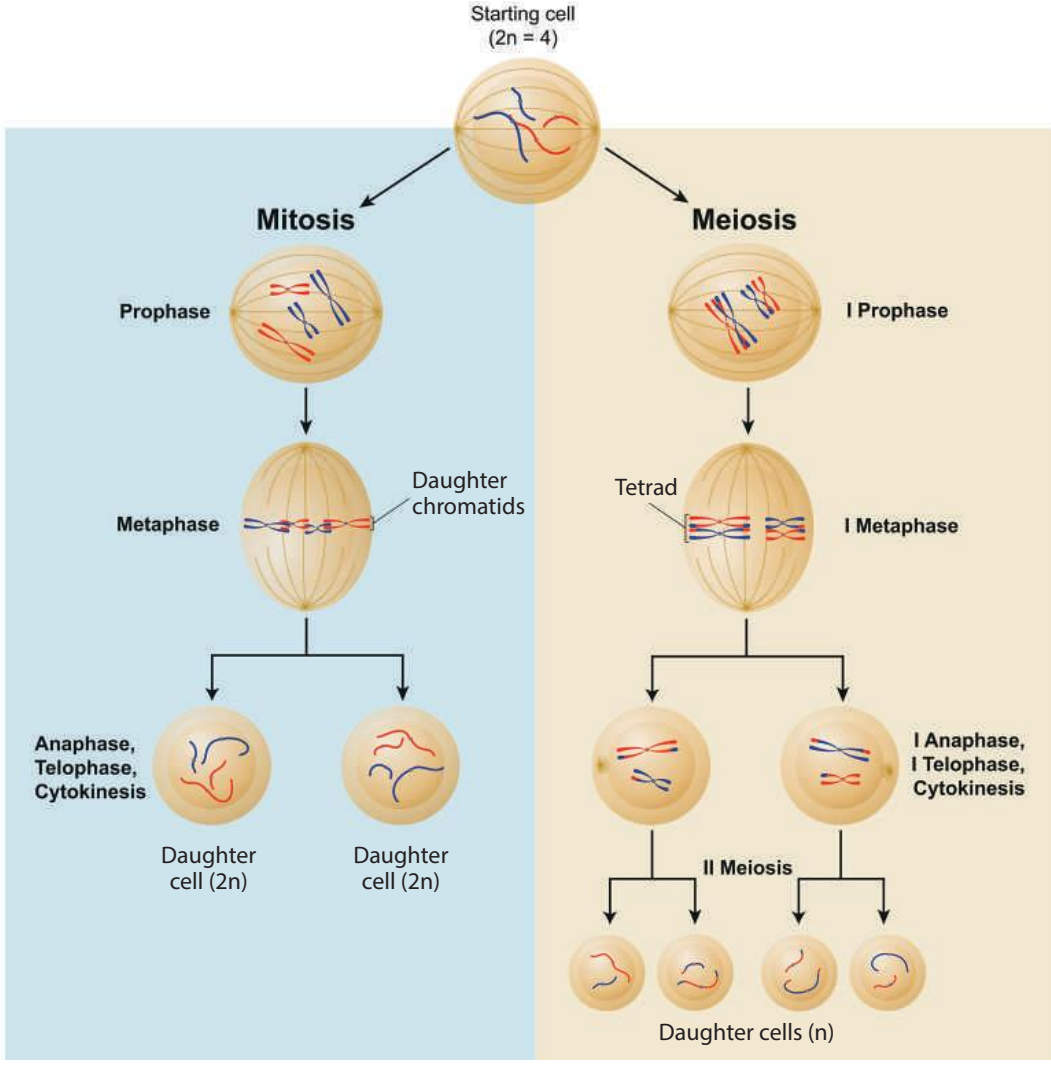


Figure 18.8: Comparison between mitosis and meiosis

Crossing over is a process that occurs during prophase 1 of meiosis when non-sister chromatids of homologous chromosomes exchange segments. This results in new combinations of genes in the gametes that are not found in either parent. Crossing over is an important cause of the genetic variation seen among offspring.

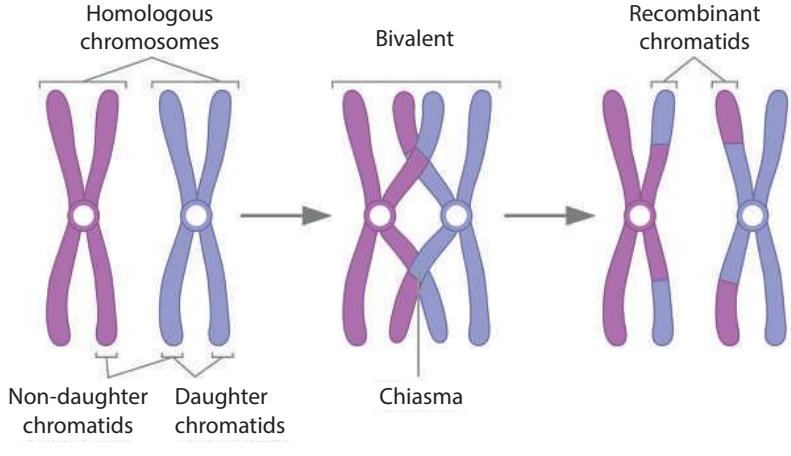


Figure 18.9: Crossing over between homologous chromosomes

CHROMOSOME MUTATION

Errors in the movement or separation of chromosomes during meiosis can cause changes that disrupt the genetic code and therefore cellular functions.

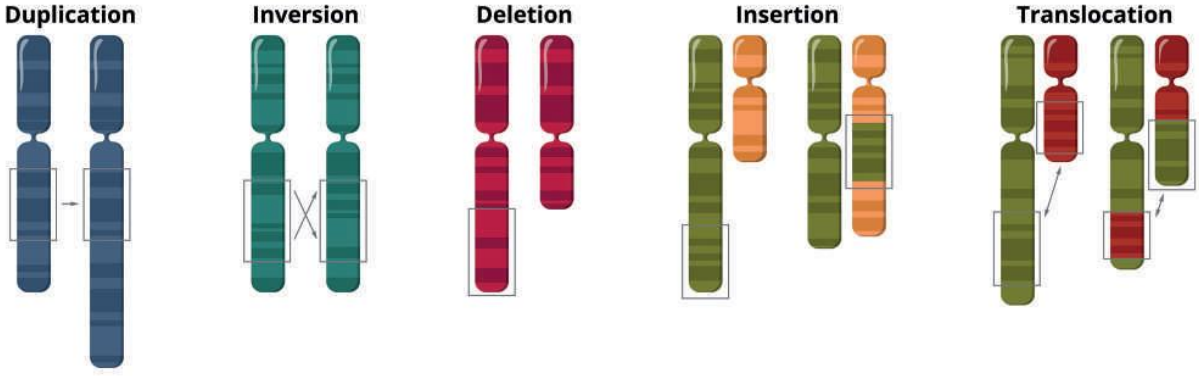


Figure 18.10: Chromosome mutations

Duplication – if sister chromatids do not split down the middle, then some genes are duplicated on one chromosome. As the sister chromatids are pulled into different cells, the cell with the duplicated genes will produce more proteins and overexpress the trait.

Inversion – a piece of the chromosome flips around and becomes reattached to the rest of the chromosome, but upside down.

Deletion – occurs when a part of the DNA is not duplicated or is lost during DNA replication.

Insertion – when part of a chromosome breaks off and attaches to the middle section of another chromosome.

Translocation – when a fragmented chromosome joins to the end of a non-homologous chromosome.

Question 1

Nearly 90% of the cell's life in the normal cell cycle is spent in which phase? (1 mark)

Question 2

In which phase of mitosis do the sister chromatids separate to move to either end of the cell? (1 mark)

Question 3

Name the structure that holds the chromatids together as a chromosome. (1 mark)

Question 4

State the name of the phase in which the chromosome become visible in meiosis. (1 mark)

Question 5

Explain why the DNA needs to be duplicated before cell division. (4 marks)

Question 6

Explain why the cells of meiosis need to have a haploid number of chromosomes whereas the daughter cells of mitosis have a diploid number of chromosomes. (2 marks)

Question 7

At which stages in mitosis are mutations most likely to occur? Why? (6 marks)

Question 8

Explain why the daughter cells of meiosis are rarely identical. (3 marks)

Question 9

Explain why mitosis needs to produce cells that are identical to the parent cells. (2 marks)

Question 10

Which of the two types of cell division is not required for the survival of an individual? Why is this type of cell division important? (2 marks)

19 DNA

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• DNA occurs bound to proteins in chromosomes in the nucleus and as unbound DNA in the mitochondria.				
• DNA stores the information for the production of proteins that determines the structure and function of cells.				
• The structural properties of the helical DNA molecule, including double-stranded, nucleotide composition and weak bonds involved in base pairing between the complementary strands, allow for its replication.				
• Discoveries made through the use of modern biotechnological techniques have increased understanding of DNA and gene expression.				

STRUCTURE

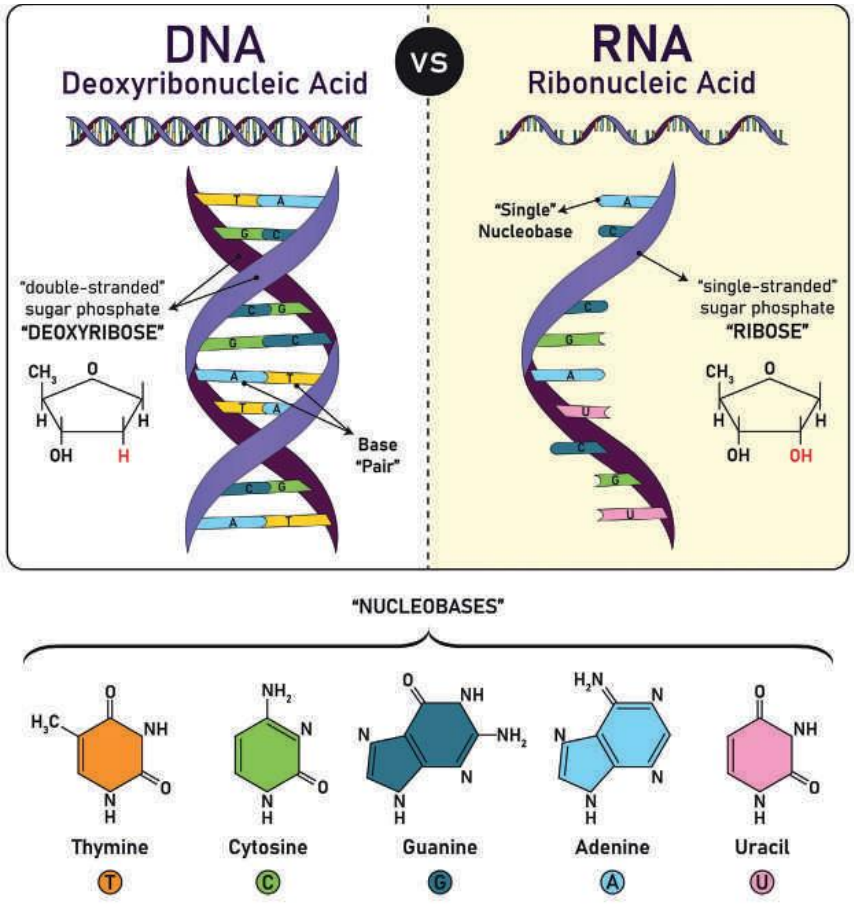


Figure 19.1: Structure of DNA

The shape of DNA is called a double helix, consisting of deoxyribose sugar and phosphate side rails and rungs of nucleotide base pairs.

The nucleotide bases are:

A – adenine

T – thymine

G – guanine

C – cytosine.

A only pairs with **T** because their structure allows for **two** hydrogen bonds to form between them.

G only pairs with **C** because their structure allows for **three** hydrogen bonds to form between them.

The bonds between the deoxyribose sugar and phosphate groups are strong covalent bonds.

Hydrogen bonds are much weaker than covalent bonds.

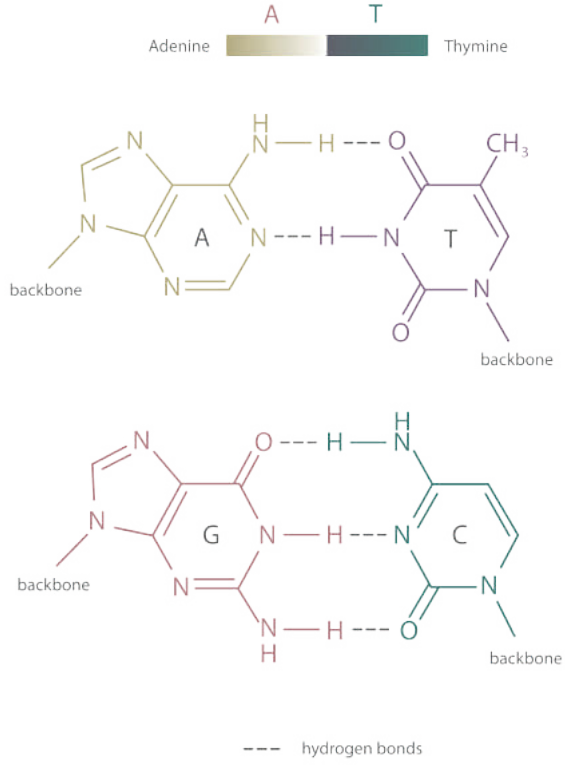


Figure 19.2: Bonding of nucleotides

NUCLEOTIDE STRUCTURE

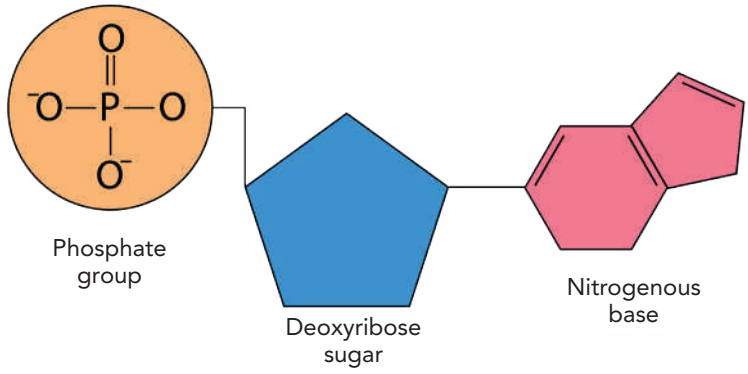


Figure 19.3: Structure of a single nucleotide

Note the location of the bonding site on the phosphate group. It is where the O^- occur, so the bonds are made to that side of the phosphate group – the 3′ end.

NUCLEOTIDE BONDING

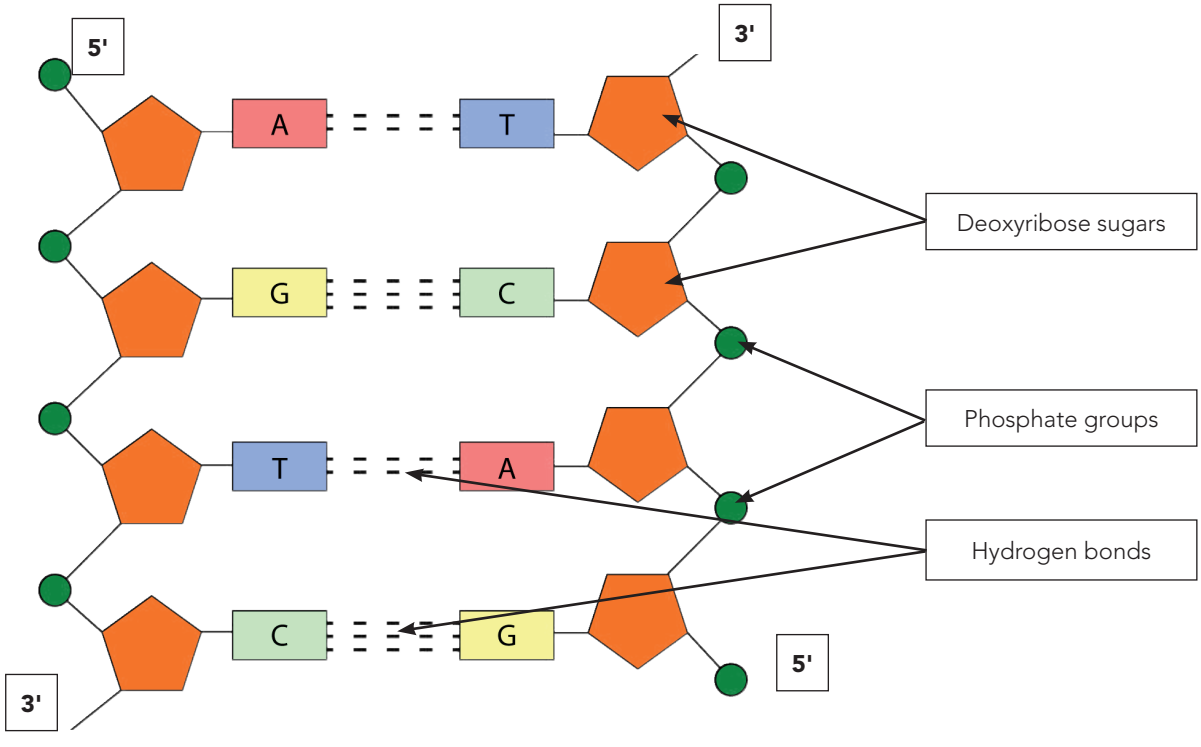


Figure 19.4: Complementary strands in DNA
From: https://www.askiitians.com/biology/biomolecules/nucleic-acid.html

Note: the orientation of the deoxyribose sugar groups in each strand – on the left they point downwards and on the right they point upwards.

The strand on the left has the top finishing with the non-binding oxygen site. These are the **5′** (five prime) ends.

The other ends that finish with the binding oxygen site are called **3′** (three prime) ends.

This is **important** because the enzyme called DNA polymerase adds nucleotides to the **3′** end only during DNA replication.

DNA CODING

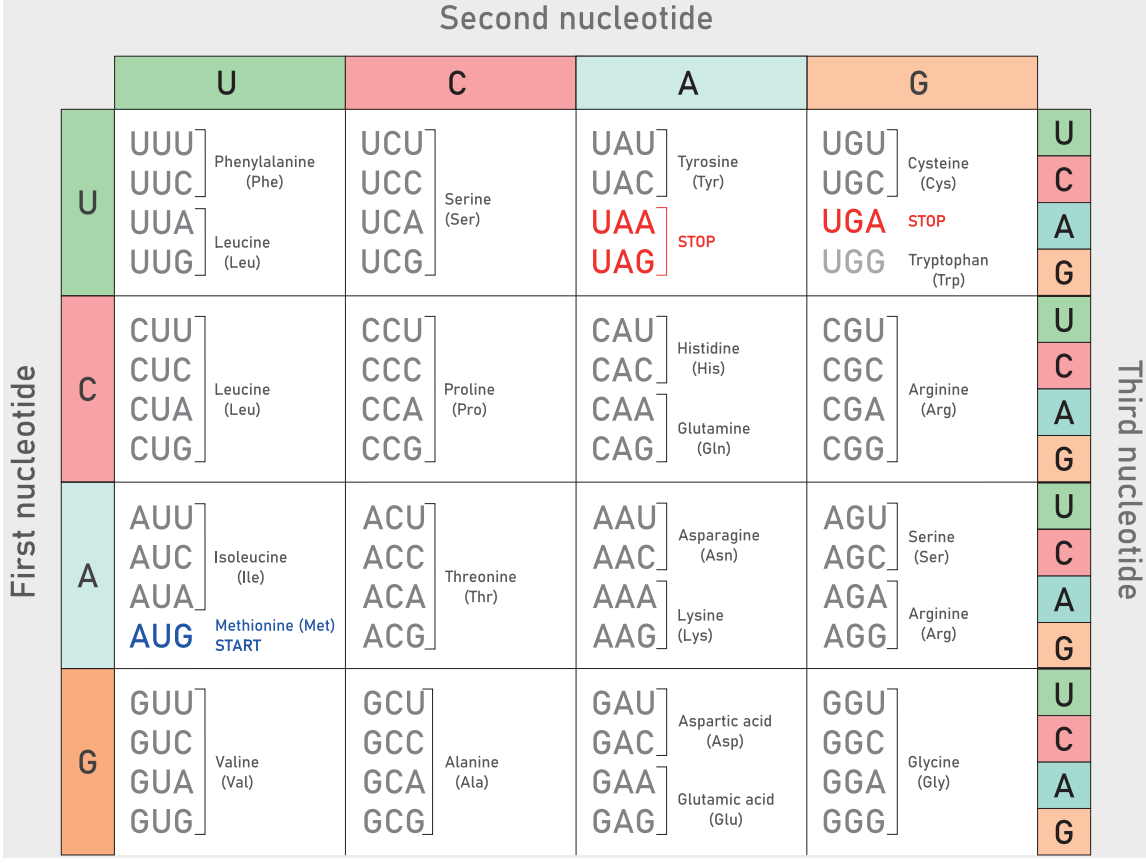


Figure 19.5: The codons related to specific amino acids

The genetic code carried on the strands of DNA is done in groups of three nucleotides called **codons**. They code for particular amino acids during protein synthesis.

The DNA code is transcribed into the sequence of bases on the mRNA.

The sequence of the bases in mRNA determines the sequence of tRNA bringing amino acids into the ribosomes for adding to the amino acid chain being produced. The codons relating to the specific amino acid are codons on the mRNA.

There are 20 different amino acids from which proteins are made.

A 2 – nucleotide code would only have 16 different variation, which is not enough to code for all twenty different amino acids.

A 3 – nucleotide (or triplet) code gives 64 different combinations of nucleotides, which is more than enough to code for all the amino acids, as well as start and stop codons. Some amino acids are coded by more than one codon.

Task 19.1: Determine the sequence of amino acids coded for by this sequence on the DNA.

GAT AAAT CT GGTCT T ATT TCC

DNA PROFILING

DNA profiling is a method of isolating and identifying variable elements within the nucleotide sequence of DNA. The technique was developed in 1984 when certain sequences of highly variable DNA (known as minisatellites) were seen to be repeated within genes. It was recognised that each individual has a unique pattern of minisatellites.

DNA profiling is used to identify individuals.

RESTRICTION FRAGMENT LENGTH POLYMORPHISM ANALYSIS (RFLP)

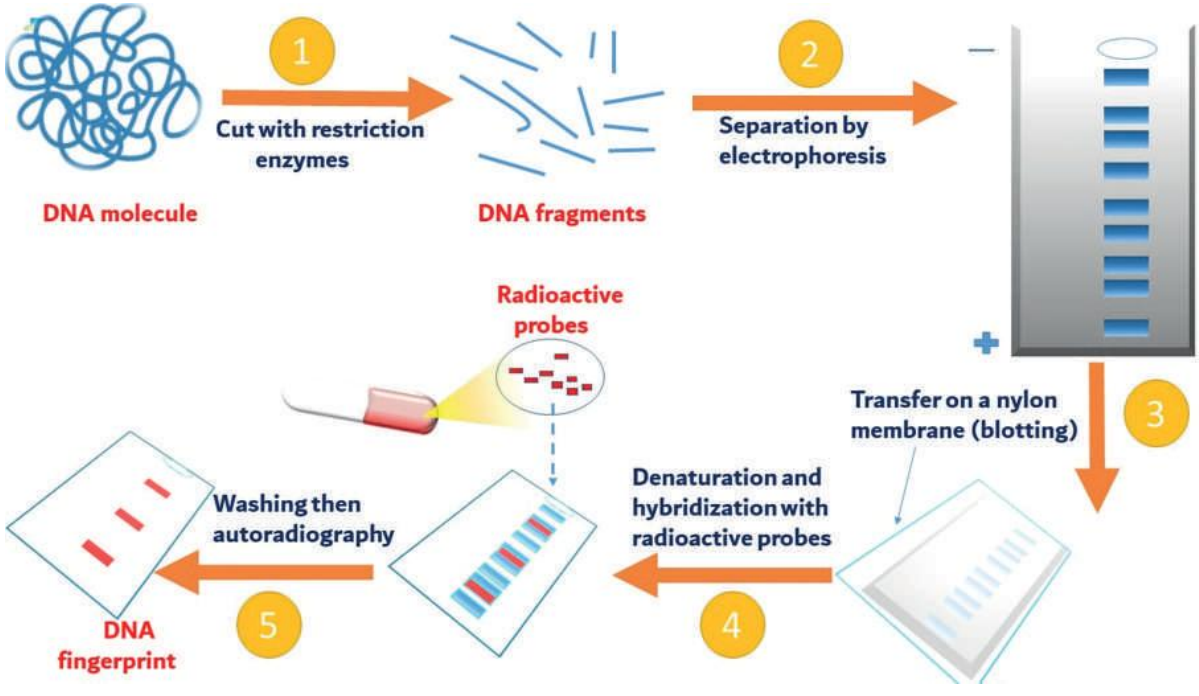


Figure 19.6: Steps in the process of DNA profiling using restriction fragment length polymorphism analysis (RFLP)

Restriction enzymes – are enzymes isolated from bacteria that cut DNA sequences at specific sequence sites, producing DNA fragments with a known sequence at each end.

HindIII, for example, is a restriction enzyme that recognises the sequence 5′AAGCTT-3′ (upper strand)/3′TTCGAA-5′ (lower strand) and cuts between the two A's on both strands.

Different restriction enzymes cut at different recognition sites i.e. different nucleotide sequences.

The DNA sample is cut into fragments of different lengths by the restriction enzymes. These fragments are separated according to their size by electrophoresis.

Electrophoresis is used to separate the DNA fragments in a gel which is subject to a relatively uniform electric field.

Fragments are separated due to their size. **Smaller fragments move through the gel faster** than larger fragments. The bands further from the placement site will be made up of smaller DNA fragments than those that haven't moved as far.

DNA fragments move towards the **positive** end of the gel because they are slightly **negatively charged**. The fragments form bands of colour in the gel. The location of the bands gives a distinctive pattern unique to individuals. The more lines on the pattern that match up the more likely the samples came from the same person. The resultant patterns of lines is called an electropherogram.

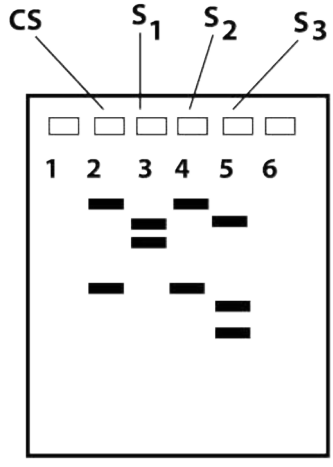


Figure 19.7: Electropherogram produced from a DNA samples using RPLF.

Task 19.2: Which suspect is most likely the culprit in the crime as result of analysing the RPLF profile?

Lane 2: Crime scene sample
Lane 3–5: Suspects 1, 2 and 3
If the selected person is not the culprit, how else could that person's DNA sample be at the crime scene?

SHORT TANDEM REPEAT (STR) PROFILES

STR analysis is a common method used to compare repeats at specific loci in DNA between two or more samples.

An STR is a microsatellite with repeat units that are 2 to 7 base pairs in length, with the number of repeats varying among individuals, making STRs effective for human identification purposes.

This method differs from (RFLP) since STR analysis does not cut the DNA with restriction enzymes. Instead, **polymerase chain reaction** (PCR) is used to discover the lengths of the short tandem repeats based on the length of the PCR products.

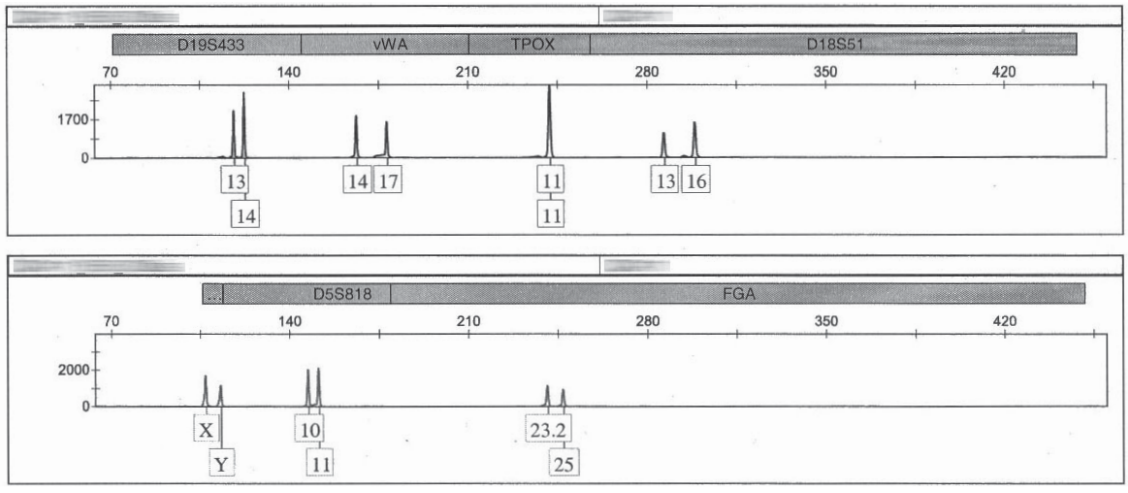


Figure 19.8: STR profile.The peaks represent DNA fragments separated by electrophoresis.

Anywhere between eight and thirteen markers spread over different chromosomes, can be used to compare samples.

The greater the number of markers used, the greater the certainty of a match being made between samples.

The data from the profiles can be stated as a set of numbers as shown below.

Table 19.1: STR marker alleles detected in samples using different microsatellite locations

Sample	Amelogenin	D3S1358	vWA	FGA	D8S1179	D21S11	D18S51
Victim	XY	14, 15	18, 20	24	13, 16	28, 30.2	14, 15
Suspect	XY	14, 15	15, 18	21, 22	13, 14	30	14, 15
Blood Stain from Crime Scene	XY	14, 15	15, 18	21, 22	13,14	30	14, 15

Sample	D5S818	D13S317	D7S820	D16S539	THO1	TPOX	CSF1PO
Victim	10, 11	8, 11	8, 11	9, 11	7, 9	9, 11	10, 12
Suspect	13	11	10	9, 12	6, 9	8, 11	9, 12
Blood Stain from Crime Scene	13	11	10	9, 12	6, 9	8, 11	9, 12

Fourteen different markers have been used to produce this STR profile. This form of recording the data from the DNA STR profile makes storage and comparisons much easier than when using the RPLF method which uses images of the bands on the gel. STR profiling is much more accurate as the bands on the RPLF gels are prone to diffusion, producing blurred edges and overlapping of bands.

Task 19.3: Use the information from Table 19.1 to determine if the suspect was the source of the blood stain at the crime scene.

DNA SEQUENCING

DNA can be sequenced by automated DNA sequencing methods and computer software to assemble the data to produce electropherograms, as below, showing the sequences of nucleotides in the DNA strand.

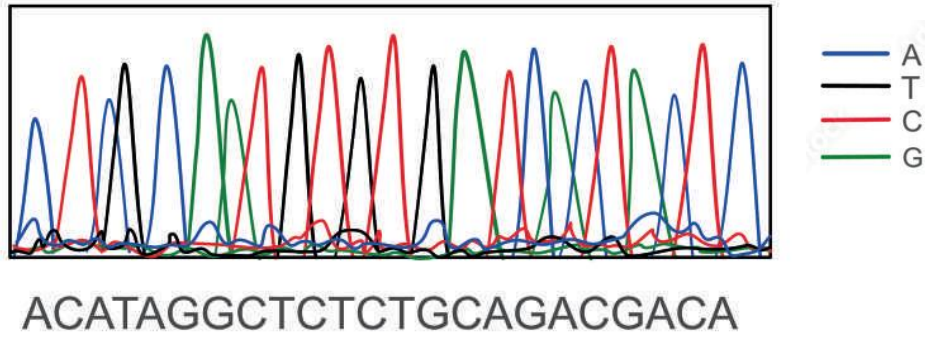


Figure 19.9: Results of automated DNA sequencing

DNA sequences can be compared for any differences between normal cells and cells from people who have a genetic condition. For example, sickle celled anaemia as shown in Figure 19.10. One change in the sequence produces a different sequence of amino acids in the protein therefore interrupts the function of the protein in the red blood cells.

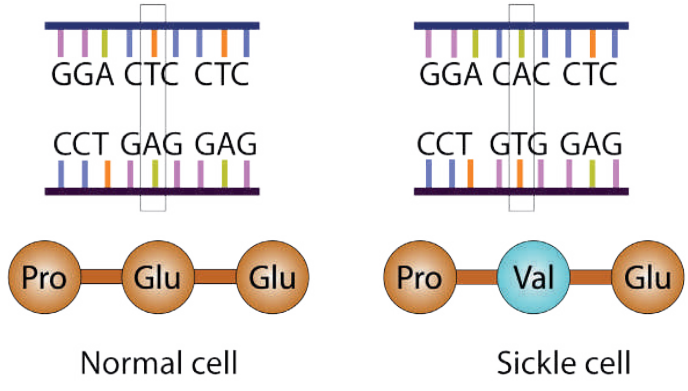


Figure 19.10: Comparison between normal cell and sickle cell DNA sequence

DNA profiling – identification of an individual

DNA sequencing – used for comparing the genome sequences of different individuals as well as different populations or species of organisms.

Question 1

Describe the factors than can influence the separation of DNA fragments in gel electrophoresis. (4 marks)

Question 2

In STR electropherogram, explain why are there sometimes one peak or number and sometimes two peaks or numbers at the same loci. (2 marks)

Question 3

GGCTCTGGATCGCTAG | GGCTCTGGATCGCTAG | GGCTCTGGATCGCTAG

The DNA code above repeats 3 times. What is this called? (1 mark)

Question 4

Name the molecule that cuts a DNA molecule at a specific base sequence. (1 mark)

Question 5

Write the complementary strand for 5′-AGCGATGTACGC-3′. (1 mark)

Question 6

Explain the importance of the hydrogen bonds between base pairs. (2 marks)

Question 7

Explain why STR profiling is used more often than RFLP profiling. (8 marks)

Question 8

What are minisatellites and why are they useful in DNA profiling? (4 marks)

Question 9

Explain the importance of the 3′ and 5′ ends of the DNA strands. (2 marks)

Question 10

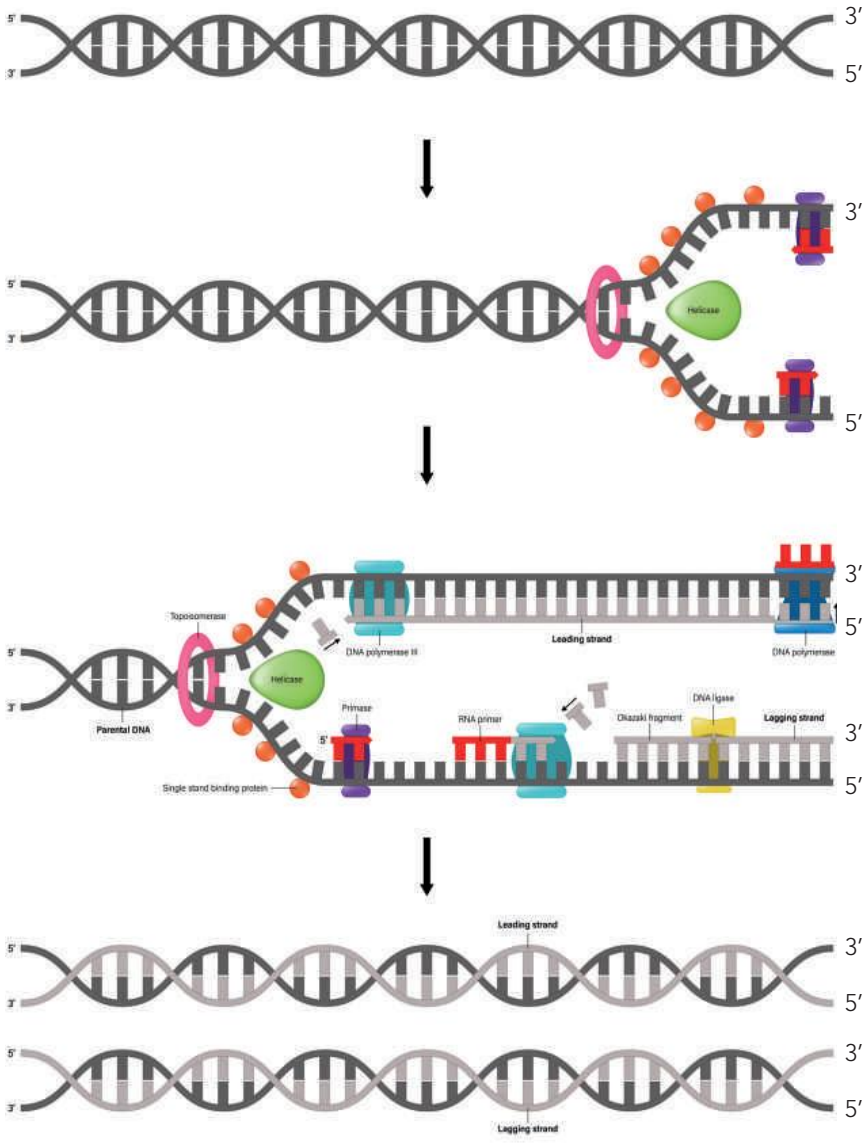
Explain why the electric charge on the DNA fragments is important in electrophoresis. (3 marks)

20 DNA REPLICATION AND MUTATION

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The structural properties of the helical DNA molecule, including double-stranded, nucleotide composition and weak bonds involved in base pairing between the complementary strands, allow for its replication.				
• Discoveries made through the use of modern biotechnological techniques have increased understanding of DNA and gene expression.				
• Crossing over, non-disjunction and random assortment of chromosomes during meiosis will produce gametes with different genetic content.				

The inheritance of characteristics and the production of proteins by cells was not fully understood until the structure of DNA was known.

Replication of DNA is semi-conservative – every double helix in the new generation consists of one complete "old" strand and one complete "new" strand wrapped around each other.



Helicase (green) acts to separate the DNA strands.

Primers (red) attach to DNA strands to indicate the start of the new DNA strand.

DNA polymerase (blue) catalyses the addition of nucleotides to the new DNA strand

Ozarki fragments form on the 5′ end strand. These are joined together by DNA ligase.

Replication is complete. All enzymes are removed and the double helix recoils.

Figure 20.1: Steps in DNA replication

Step 1: 'Unzipping'

DNA helicase disrupts the hydrogen bonds between the base pairs.

One strand is oriented in the 3′ to 5′ direction (leading strand) while the other is oriented 5′ to 3′ (lagging strand). The two sides are therefore replicated with two different processes to accommodate the directional difference. (Nucleotides are only added to the 3′ end of the strand.)

Step 2: Primer Binding

The leading strand is the simplest to replicate. Once the DNA strands have been separated, a **primer** (a short piece of RNA) binds to the 3′ end of the strand. The primer always binds at the starting point for replication.

Step 3: Elongation

DNA polymerases are responsible adding nucleotides to the strand by a process called elongation. Because replication proceeds in the 3′ to 5′ direction on the leading strand, the newly formed strand is continuous.

The lagging strand begins replication by binding with multiple primers. Each primer is only several bases apart. DNA polymerase then adds nucleotides in groups, called **Okazaki fragments**, to the strand between primers. This process of replication is discontinuous as the newly created fragments are disjointed.

Step 4: Termination

An enzyme called **DNA ligase** joins Okazaki fragments together forming a single unified strand. The ends of the parent strands consist of repeated DNA sequences called **telomeres**. Telomeres act as protective caps at the end of chromosomes to prevent nearby chromosomes from fusing.

In the end, replication produces two DNA molecules, each with one strand from the parent molecule and one new strand.

This process of replication does not always occur perfectly. Errors can occur causing mutations.

GENE MUTATION

Once the coding method used to translate the information in the genetic code and how it was used to produce specific proteins was known, researchers could then understand the basis of gene mutations. Changes in the DNA sequence caused differences in proteins which controlled cellular functioning.

Substitution – one nucleotide is changed to another caused by an error in the pairing during replication.

Deletion – one or more nucleotides are removed from the sequence

Insertion – one or more nucleotides are added to the sequence

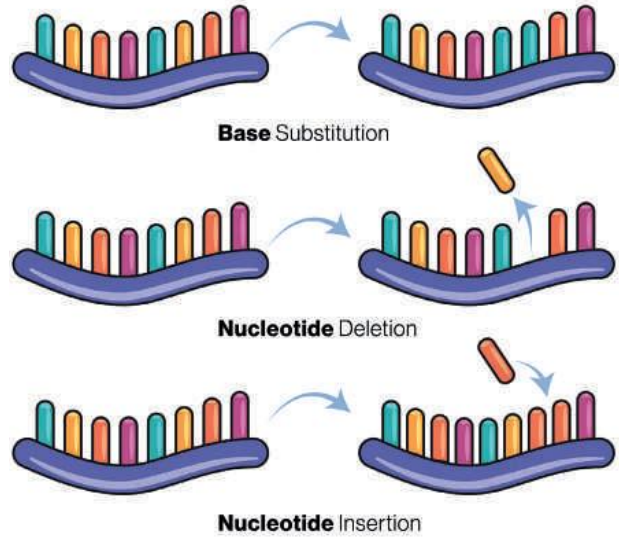


Figure 20.2: DNA mutation types

Sickle cell anaemia is caused by a single change in the genetic code of 576 – nucleotide long gene for haemoglobin. This change causes a change in the sequence of amino acids in the haemoglobin chain which disrupts its function in red blood cells. See Chapter 19, Figure 19.10.

Question 1

Where do the nucleotides used in replication come from? (1 mark)

Question 2

Explain how Ozarki fragments are formed. (2 marks)

Question 3

Describe how each type of gene mutation could be caused in the processes of replication of DNA. (6 marks)

Question 4

Explain why it is important to replicate the nucleotide sequence as closely as possible. (2 marks)

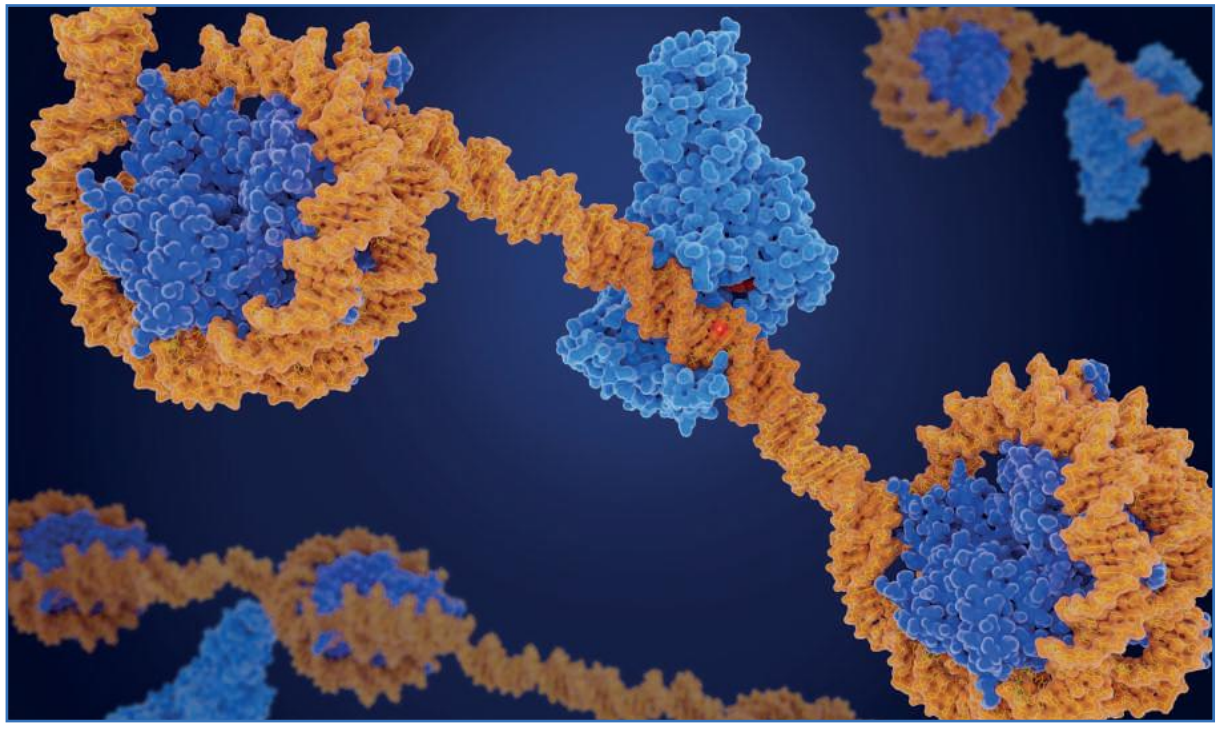
Question 5

Explain why some gene mutations do not cause any change in the phenotype. (2 marks)

Question 6

Outline the consequences of a gene mutation in the genetic code for DNA ligase. (2 marks)

21 EPIGENETICS



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Epigenetics is the study of phenotypic expression of genes, which depends on the factors controlling transcription and translation during protein synthesis, the products of other genes, and the environment.				

When the cell divides, the DNA becomes tightly coiled, winds around histone proteins then coiled again into the visible chromosomes observed during cell division.

DNA is the **genome** but the histone and other chemicals are called the **epigenome**.

In general, epigenetic marks are reprogrammed between generations in order that the cells of the pre-implantation embryo are totipotent (have the capacity to develop into all cell types).

Epigenetic markers **control when genes can be accessed for transcription**.

Almost all cells in the body have the same genome, but some become blood cells and others become bone or muscle cells. This is associated with changes in the particular subset of genes that are transcriptionally active and is the result of epigenetic changes.

Epigenetics is defined as mitotically heritable changes in **gene activity and expression** that occur without the alteration of the genetic sequence (genome).

Epigenetics is an additional layer of instructions that controls how our DNA is interpreted – how our genes are controlled and expressed.

Epigenetic mechanisms change the way genes are packaged in the cell nucleus, and involve changes in chemical groups that can attach to DNA, or changes in the way RNA molecules interact with our DNA.

Genetic changes can alter which protein is made; epigenetic changes affect gene expression to turn genes "on" and "off."

METHYLATION OF DNA

This is the addition or removal of a methyl group (CH_3). Adding a methyl group is associated with transcriptional repression – **gene silencing**.

DNA methylation is a normal part of embryonic cell differentiation and its influence persists through the life of the cell and can be passed on during cell division.

The **histone proteins** associated with DNA can also be changed by environmental factors. This causes the DNA to be more tightly or loosely bound to the histones, making them more or less available for transcription.

The changes produced by **methylation** and **histone modification** are localised to the particular cells in which it occurs. If, for example, the melanin production gene was silenced in a stem cell of the dermis of the skin, it would show as a white patch on the skin. If it occurred in the epidermal cell that is about to be removed by naturally sloughing of skin cells, then there would be no observable effect.

Methylation is affected by the following:

- diet
- stress
- heavy metals
- pesticides
- diesel exhaust
- tobacco smoke.

Task 21.1: Watch the following short video

Epigenetics Amoeba sisters https://youtu.be/MD3Fc0XOjWk.
Write a succinct list of points to summarise the content.

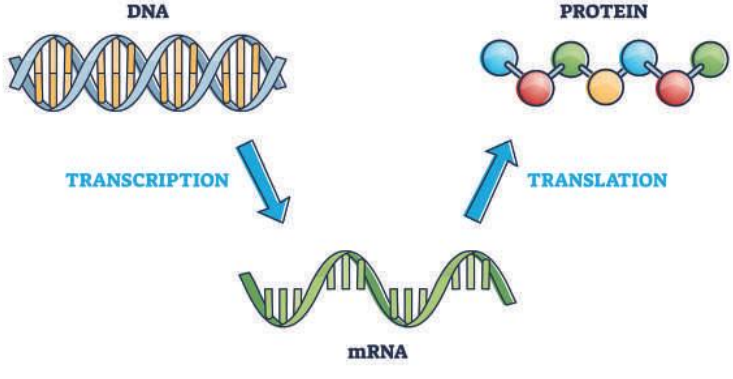


Figure 21.1: Steps in gene expression

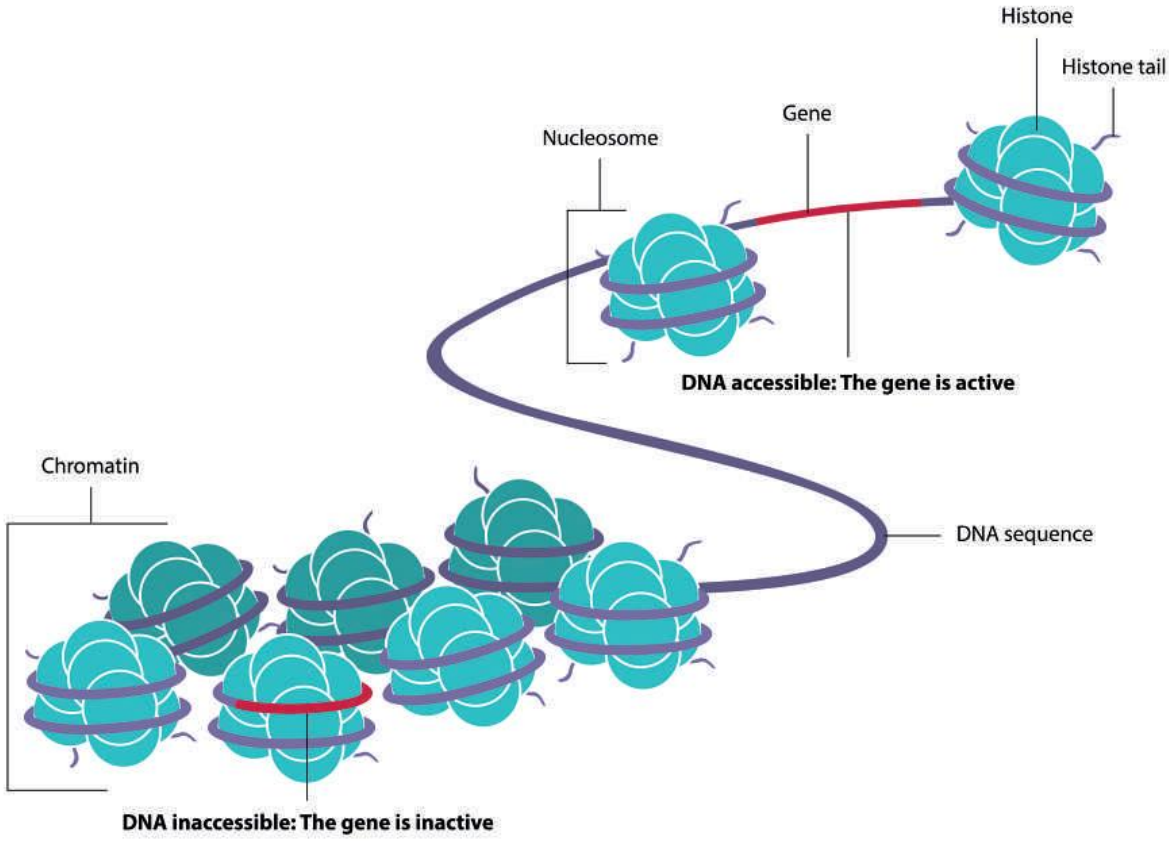


Figure 21.2: Control of gene transcription

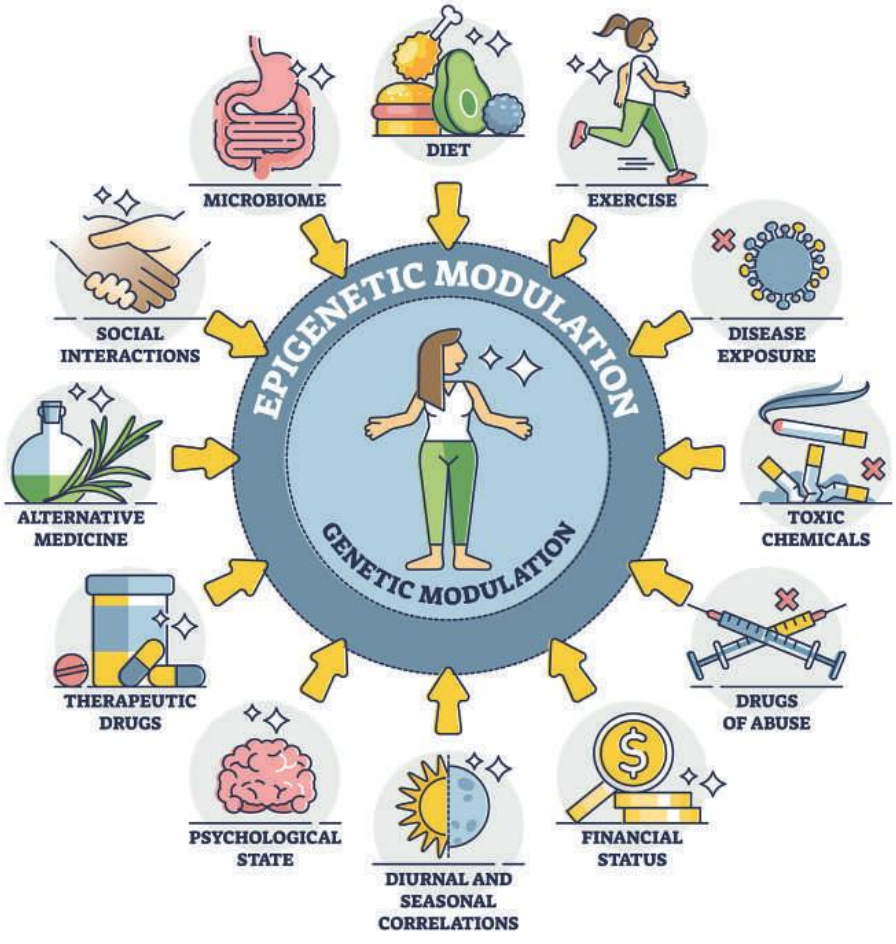


Figure 21.3: Factors affecting gene expression

Question 1

Use your understanding of epigenetics to explain the following situations.

(a) Identical twins have different rates of incidence of cancer. (3 marks)

(b) Some smokers develop lung cancer after smoking cigarettes for a very short time, while some long-term smokers never develop lung cancer. (2 marks)

(c) More cancers occur in older people. (2 marks)

(d) Why women planning to get pregnant soon or are in the early stages of pregnancy should be conscious of their diet and lifestyle. (2 marks)

Question 2

Why do identical twins become less identical as they grow older? (1 mark)

PROTEIN SYNTHESIS

22

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Protein synthesis involves the transcription of a gene on DNA into messenger RNA in the nucleus, and translation into an amino acid sequence at the ribosome with the aid of transfer RNA.				

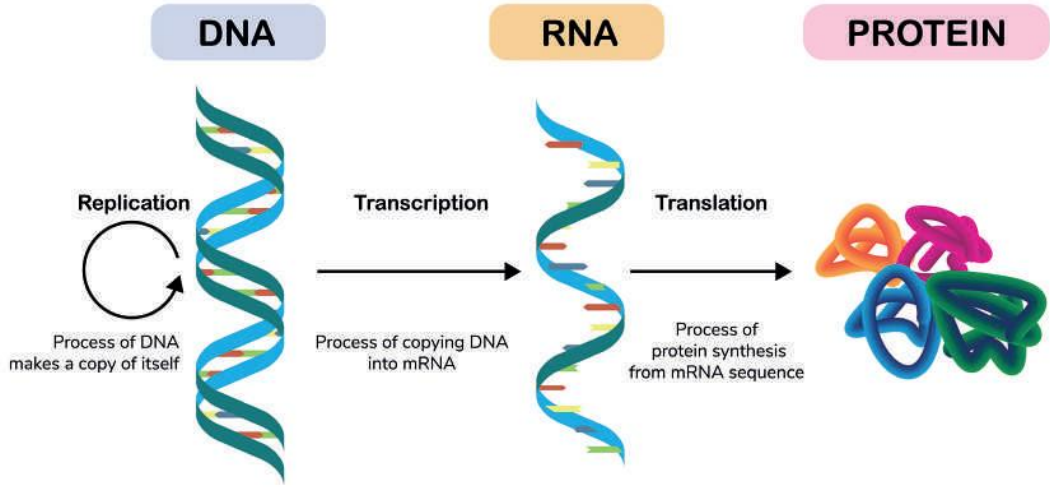


Figure 22.1: Central dogma for protein synthesis

Proteins are produced in cells under instruction from the nucleus based on the genetic code of the DNA.

DNA does not leave the nucleus. RNA is the connection between the DNA and the centres of proteins production, the **ribosomes**.

Messenger RNA (mRNA) transcribes the nucleotide sequence from the DNA and takes it to the ribosomes in the cytoplasm or attached to the endoplasmic reticulum. mRNA does not have thymine bases, but uses **uracil** instead. **A** on DNA joins to **U** in mRNA.

Not all the sequence of nucleotides **transcribed** from the nuclear DNA is useful for **translation** into proteins.

Introns – these are 'junk' sequences; parts of the DNA not used in the sequence for protein production, but may have control functions on regulating gene activity.

Exons – these are functional sequences for the production of proteins.

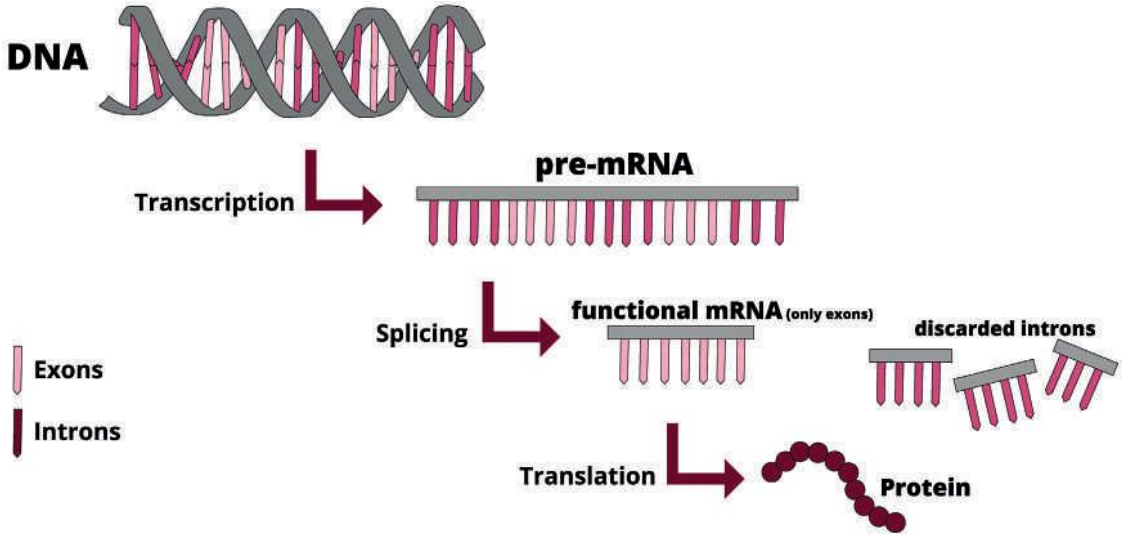


Figure 22.2: Introns and exons

Splicing occurs to remove the introns leaving only the nucleotide sequence that will code for the amino acids in the protein chain.

PROTEIN SYNTHESIS

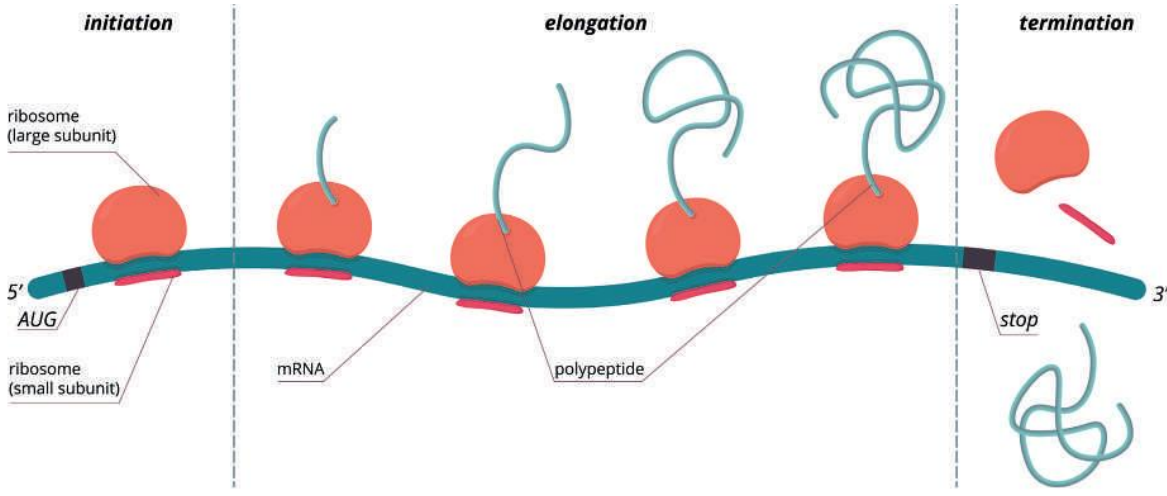


Figure 22.3: Overview of protein production at the ribosome using functional mRNA

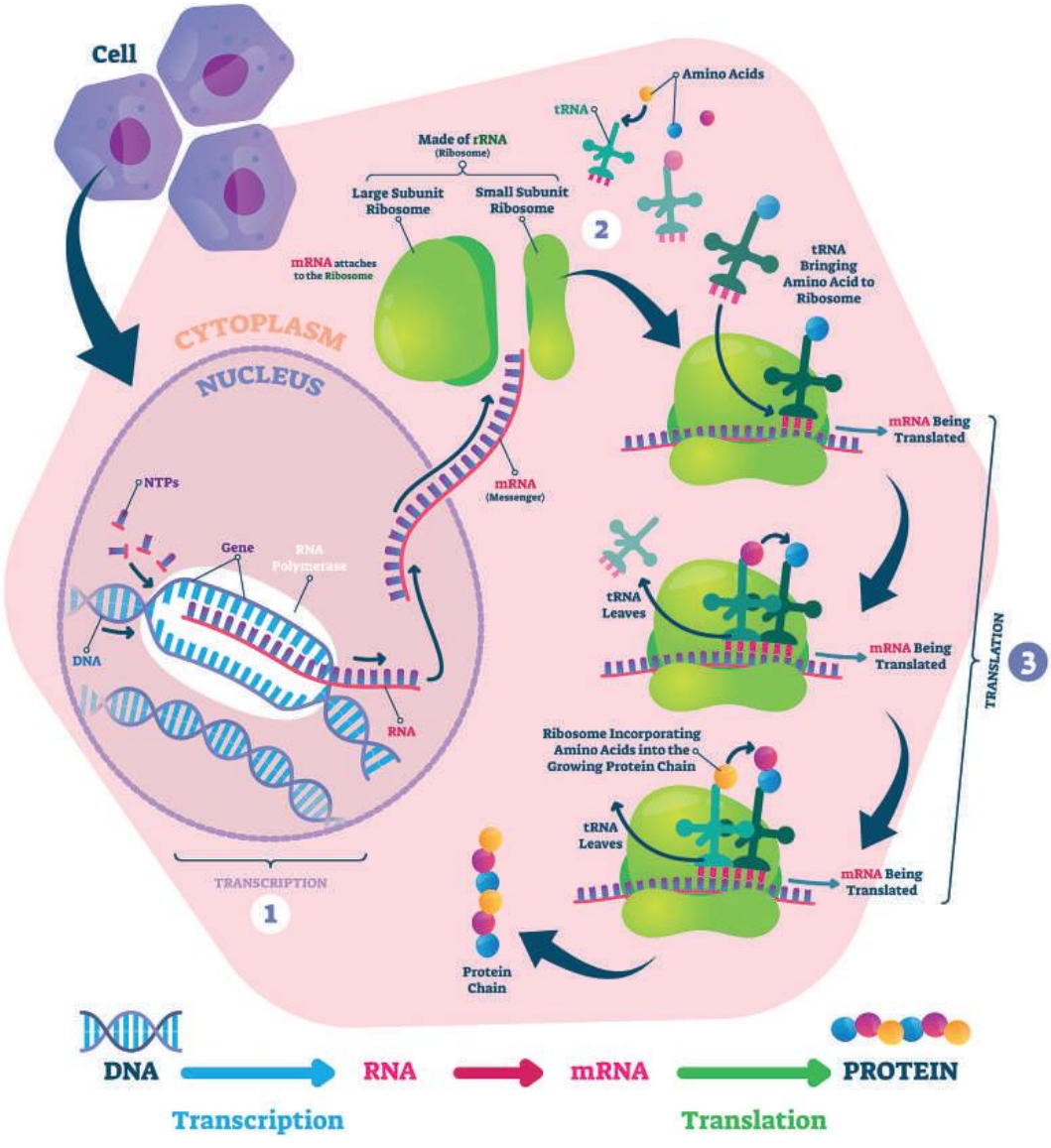


Figure 22.4: Steps in protein synthesis

Transfer RNA (tRNA) is found in the cytoplasm. Each tRNA has a **triplet code for a specific amino acid**. It attaches to the amino acid and takes it to the ribosome ready to be placed in the sequence when it's code comes up.

Table 22.1: Steps in the production of a protein at the ribosome

Processes	Description
Transcription	Process of producing an equivalent RNA copy of a sequence of **DNA**, and introns are removed leaving only the exons.
Translation – initiation	Messenger RNA (mRNA) produced by transcription is decoded by the ribosome to produce a specific amino acid chain. Transfer RNA (tRNA) starts searching the mRNA for the start codon.
– elongation	The elongation of the polypeptide chain begins with the tRNA entering the ribosome. • The tRNA carrying its amino acid is correctly oriented in the ribosome. • Formation of the peptide bond between the amino acids occurs. • tRNA and amino acid part and tRNA returns to the cytoplasm to find another complementary amino acid. • After every peptide bond formation, the mitochondrion shifts by one codon along the mRNA.
– termination	It is the final phase of the translation process. If any of the termination codon enters the ribosome, the translation process stops. There is no tRNA to add an amino acid to the chain. The protein formed as a result of this whole process, is released from the ribosome and the translation process ends.

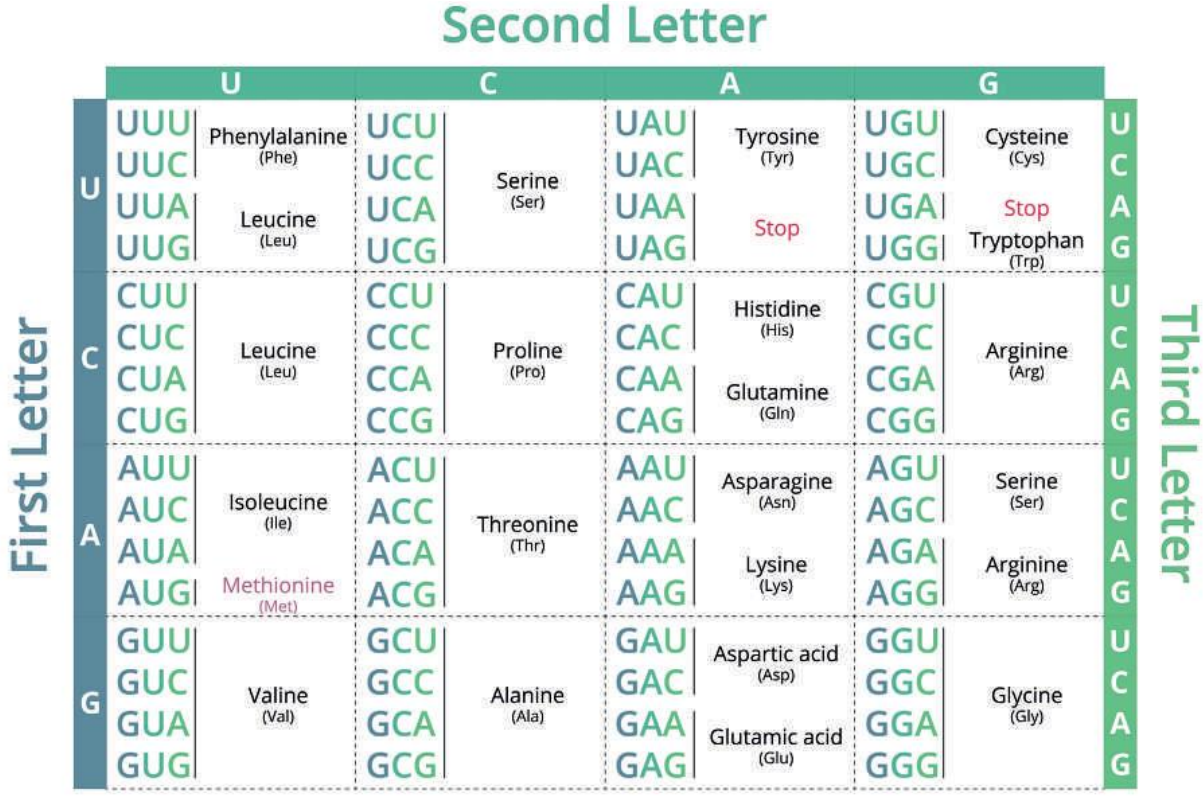


Figure 22.5: Triplet codons in RNA and corresponding amino acids

FINAL PROTEIN STRUCTURE

The function of the protein is dependent on the protein's shape.

After the amino acid chain has been produced at the ribosome, it moves into the Golgi body where it can be modified in different ways.

Primary structure – sequence of amino acids in the chain comes from the ribosome to the Golgi body

Secondary structure – the orientation of the chains of amino acids into helices or flat sheets

Tertiary structure – the 3D shape and arrangement of helices and sheets with twists and folds

Quaternary structure – combinations of different protein chains to form one functional protein e.g. haemoglobin has 2 alpha and 2 beta chains surrounding an iron atom.

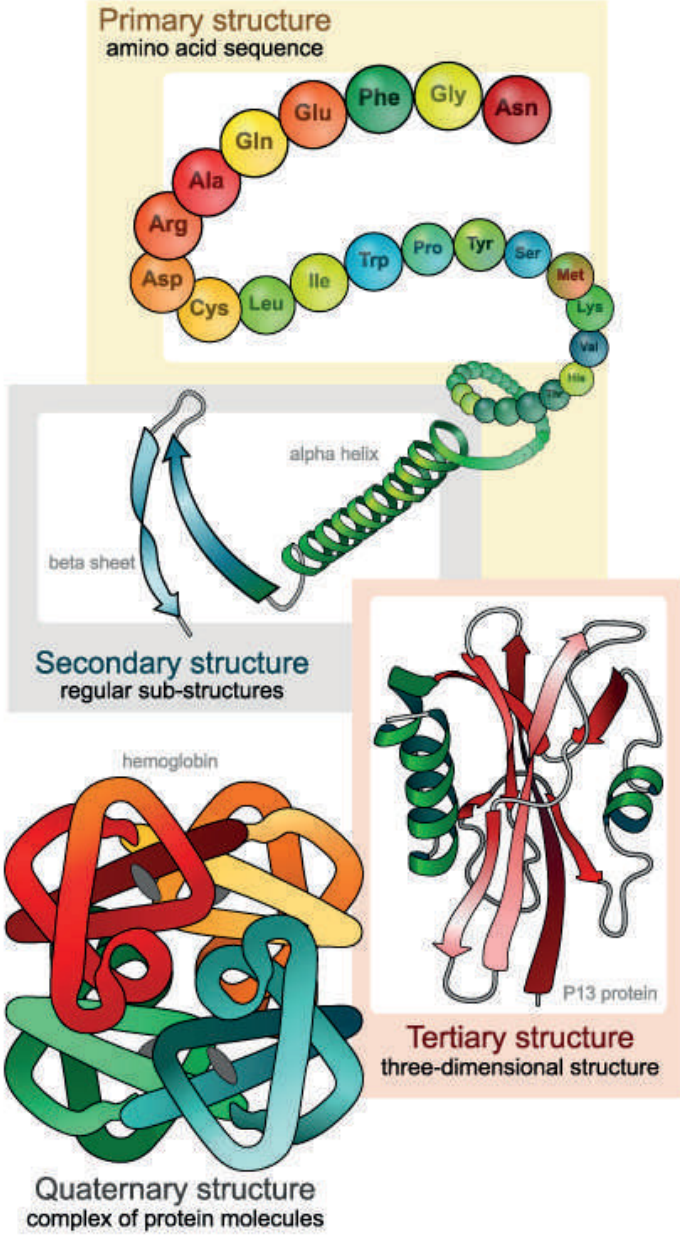


Figure 22.6: Protein structure. The coloured balls at the top of this diagram represent different amino acids. Amino acids are the subunits that are joined together by the ribosome to form a protein. This chain of amino acids then folds to form a complex 3D structure.
From: LadyofHats, Public domain, via Wikimedia Commons

Question 1

Explain why the sequence of amino acids in the protein is important to its function. (2 marks)

Question 2

Compare the code on the gene in the nucleus with the code on the mRNA that transcribes it. (1 mark)

Question 3

In 1966, Marshall Nirenberg and Gobind Khorana made synthetic RNA of all uracil nucleotides. The protein produced using this RNA was made up of only the amino acid phenylalanine. How could this technique be used to link the genetic code to the amino acid sequence in proteins? (1 mark)

Question 4

Explain how STOP codons work. (3 marks)

Question 5

Write the amino acid sequence for the following RNA code. (3 marks)

AUGCCUGGUCAUGUACUACAACUUCAUUCUUUAAAGUCUUAA

Question 6

What is the DNA code for the protein in question 5? (3 marks)

Question 7

Determine the consequences of change in the sequence when the third G is deleted through a mutation of the DNA. (6 marks)

Question 8

In sickle cell anaemia, the amino acid glutamic acid is changed to the amino acid valine in the chain of amino acids that produce the alpha chains of haemoglobin. How does this impact on the quaternary structure of haemoglobin? (3 marks)

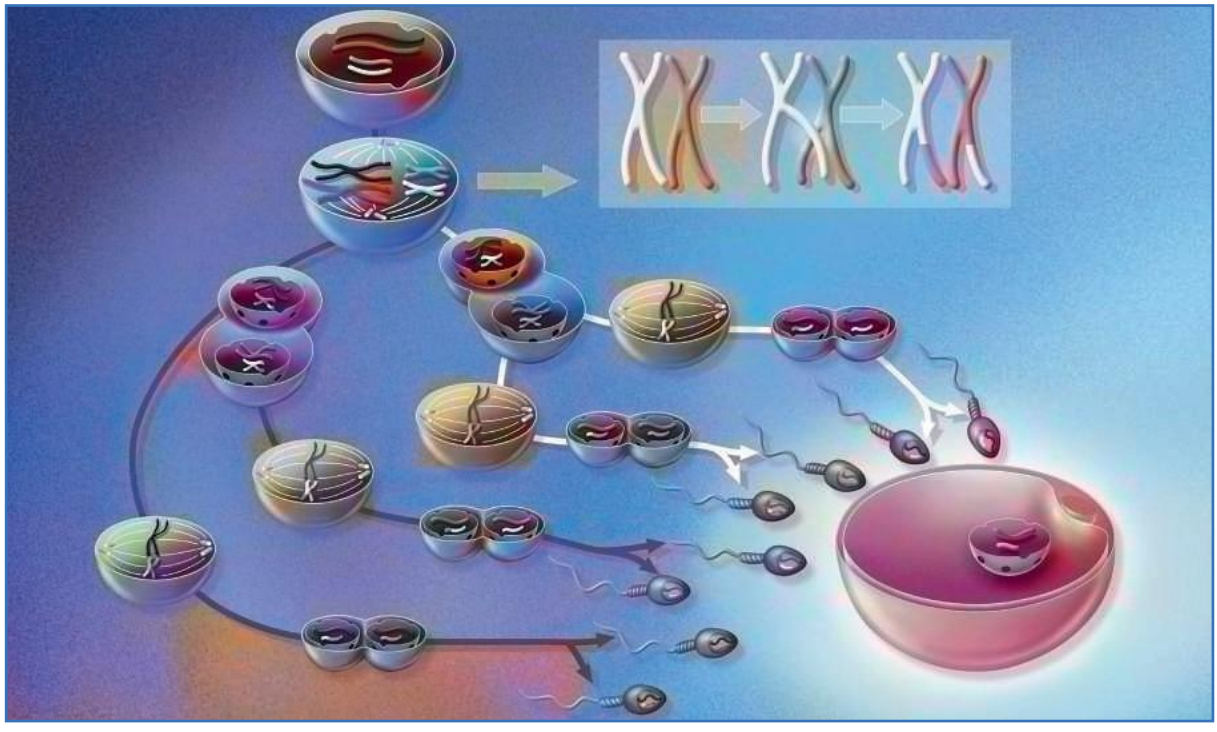
Question 9

Explain why the overall structure of a protein is important. (2 marks)

Question 10

Explain why the STRs used in DNA profiling are found in introns. (2 marks)

23 GAMETE FORMATION



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Human gametes are produced through spermatogenesis and oogenesis, which are specific forms of meiosis, but vary significantly in process and products.				

Sexual reproduction requires the combination of DNA from two parents.

One set of genetic information comes from the father and one from the mother.

When the genetic information combines, the offspring has two sets genetic information in each cell.

The genetic information is carried in the chromosomes.

one set of chromosomes – **haploid** number

two sets of chromosomes – **diploid** number

Human diploid number = 46

Human haploid number = 23

Meiosis produces daughter cells that have haploid numbers of chromosomes.

These daughter cells undergo modification to produce **gametes** – **reproductive cells**.

Males – **spermatogenesis** produces the male gamete – sperm – in the testes

Female – **oogenesis** produces the female gamete – ovum – in the ovaries

SPERMATOGENESIS

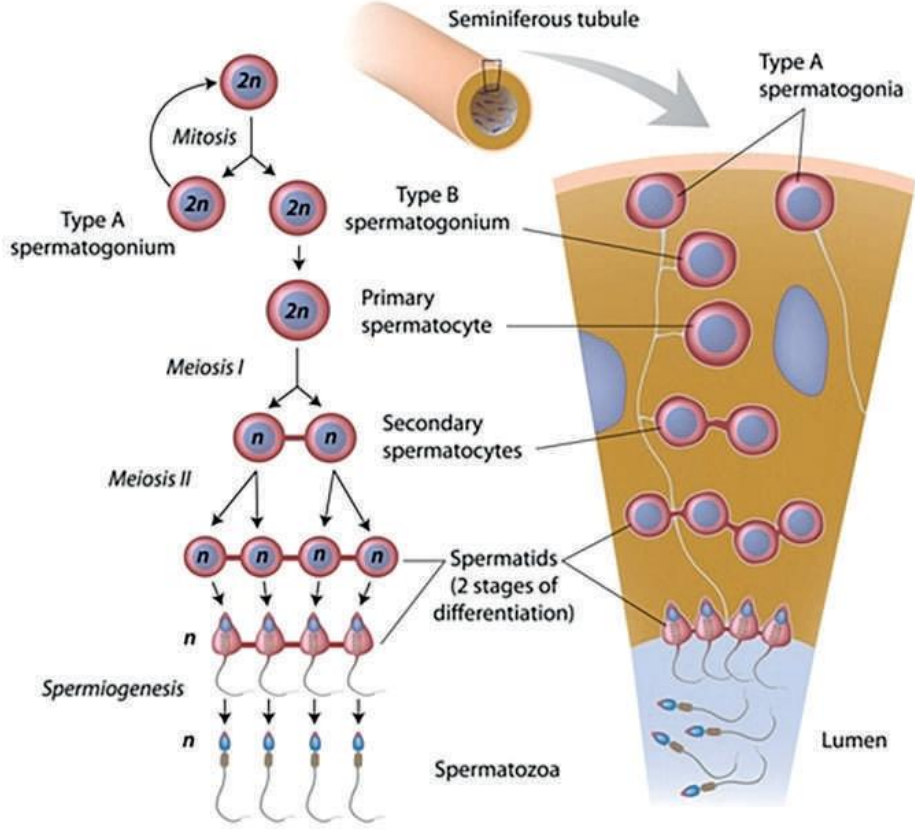


Figure 23.1: Spermatogenesis

As the cells undergo the process of meiosis, they move towards the lumen or cavity of the seminiferous tubule.

The final processes see the spermatids develop a flagellum (tail) and lose most of the cytoplasmic contents, taking on the specific structure of the sperm occurs as they move through the seminiferous tubules and final maturation occurs in the epididymis.

For humans, the entire process of spermatogenesis is variously estimated as taking about 74 days and including the transport on ductal system, it takes a total of about 3 months. Testes produce 200 to 300 million spermatozoa daily. However, only about half or 100 million of these become viable sperm.

If the sperm are not ejaculated after that time, they degenerate and are reabsorbed into the body tissues.

Although spermatogenesis continues throughout many males' lives, sperm quality tends to decrease with age.

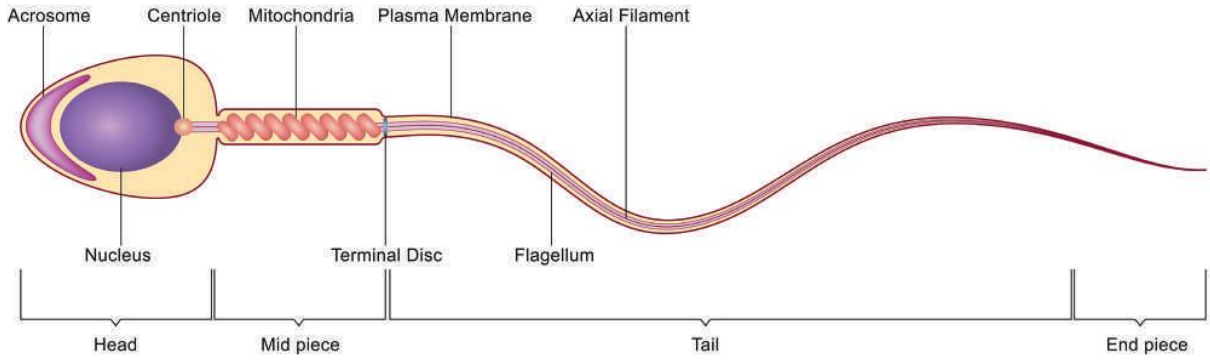


Figure 23.2: Structure of a mature sperm

Acromere – contains digestive enzymes

Nucleus – contains the genetic information

Mid piece – contains mitochondria

Tail – contains contractile filaments

OOGENESIS

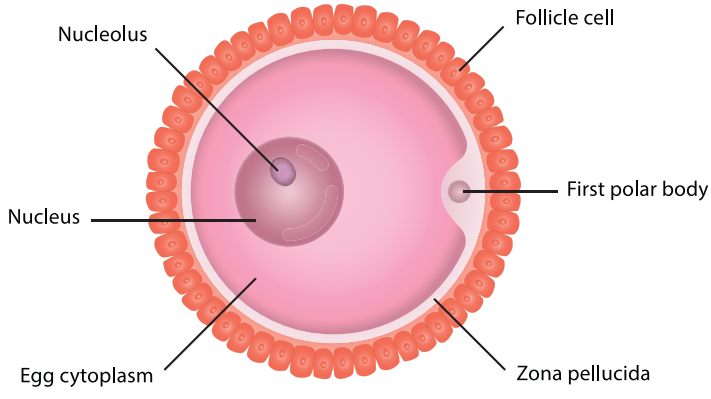


Figure 23.3: Structure of the ovum

The ovum contains a large amount of cytoplasm to provide for survival of the fertilised ovum through the fallopian tube until it is firmly implanted into the endometrium where it will have a continual source of nutrients. During this time the zygote will undergo cell division which requires a lot of energy.

In oogenesis, there is **unequal division** of the cytoplasm forming **polar bodies**. These contain the genetic materials but little else. These breakdown and are absorbed by the surrounding tissue.

Only **one** of the daughter cells of meiosis in oogenesis survives to produce an **ovum**.

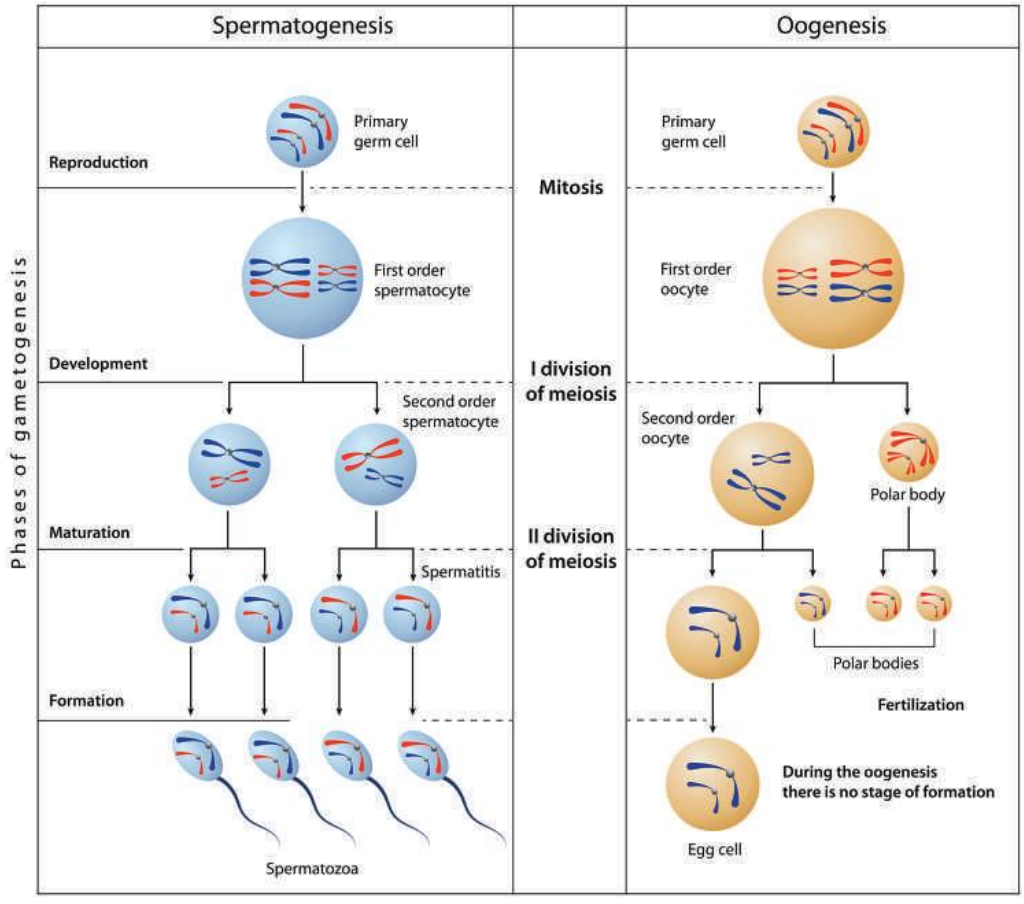


Figure 23.4: Comparison of spermatogenesis and oogenesis

The human ovum is the largest cell in the body, whereas the human sperm is amongst the smallest.

Sperm cells are so small as they only contain 23 chromosomes in a nucleus, some mitochondria, and a few other parts.

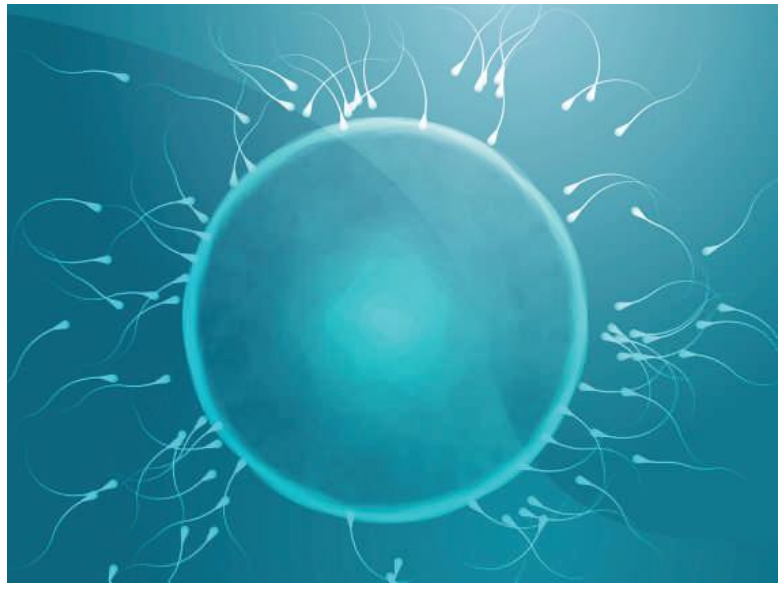


Figure 23.5: Ovum surrounded by sperm

Figure 23.5 provides an indication of the size difference between the two gametes.

The human sperm head is about 5.1 μm by 3.1 μm and a tail is about 50 μm long.

The human ovum measures approximately 120 µm in diameter.

Question 1

Explain the functions of the following structures of the sperm.

(a) Acromere (1 mark)

(b) Midpiece (1 mark)

(c) Tail (1 mark)

Question 2

Compare age of ova and sperm that produce zygote ready for implantation. (4 marks)

Question 3

Compare the size of male and female gametes. (5 marks)

Question 4

Describe what happens to the polar bodies produced during oogenesis. (1 mark)

Question 5

Explain the role of the zona pellucida surrounding the ovum. (2 marks)

Question 6

What is the corona radiata around the ovum and what is its role? (3 marks)

24 PEDIGREE CHARTS

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Probable frequencies of genotype and phenotype of offspring can be predicted using Punnett squares and by taking into consideration patterns of inheritance, including the effects of dominance, co dominance, autosomal or sex-linked alleles, and multiple alleles: Huntington's disease, phenylketonuria (PKU), ABO blood groups, red–green colour blindness/ haemophilia show different inheritance patterns.				
• Pedigree charts can be constructed for families with a particular genetic disorder and can be used to reveal patterns of inheritance and assist in determining the probability of inheriting the condition in future generations.				

In the late 1950's geneticists established a direct relationship between human genetic disorders and abnormal chromosome numbers in cells. They built on information of previous scientists.

- **1860's**: Gregor Mendel, suggested that each parent had 'factors' that were contributed to the offspring and that inheritance followed a basic set of 'laws'.
- **1905**: the combination the sex chromosomes in individuals was described by Nettie Stevens and Edmund Wilson.
- **1910**: Thomas Hunt Morgan demonstrated that some characteristics were sex-linked.
- **1910**: Reginald Punnet popularised a shorthand method to follow traits through experimental crosses.
- **1931**: Barbara McClintock and Harriet Creighton obtained cytological proof that crossing over occurs during meiosis to form new combinations of characteristics.

SCIENTIFIC CONVENTION FOR DRAWING PEDIGREE CHARTS

- the dominant allele is written as a capital letter and the recessive allele is written as a lower case letter (**not** a small capital letter).
- the letters chosen should have different forms for the capital and lower case letters to make identification easier e.g. Ee; Ff, Gg, Hh, Dd.
- difficult to differentiate letters such as Ss, Cc, Pp, Ww are to be avoided.

Setting out a demonstration of a monohybrid cross (inheritance of one characteristic)

1. State the parental genotypes

   Mother's genotype **Bb** Father's genotype **Bb**

2. State the gametes each parent can produce

   Female gametes: **B** and **b** Male gametes **B** and **b**

3. Move the gametes to the Punnet square below.

		Male's gametes	
Female's gametes		**B**	**b**
	B	**BB**	**Bb**
	b	**Bb**	**bb**

4. Fill in the shaded squares with the combination of the female and male gametes
5. State the genotypes and phenotypes of the offspring (shaded squares)

   BB – homozygous dominant 1 25%

   Bb – heterozygous 2 50%

   bb – homozygous recessive 1 25%

6. State how many of each.
7. Convert numbers of each to percentages.
8. Re-read the question to find out what answer you are to give based on this information.

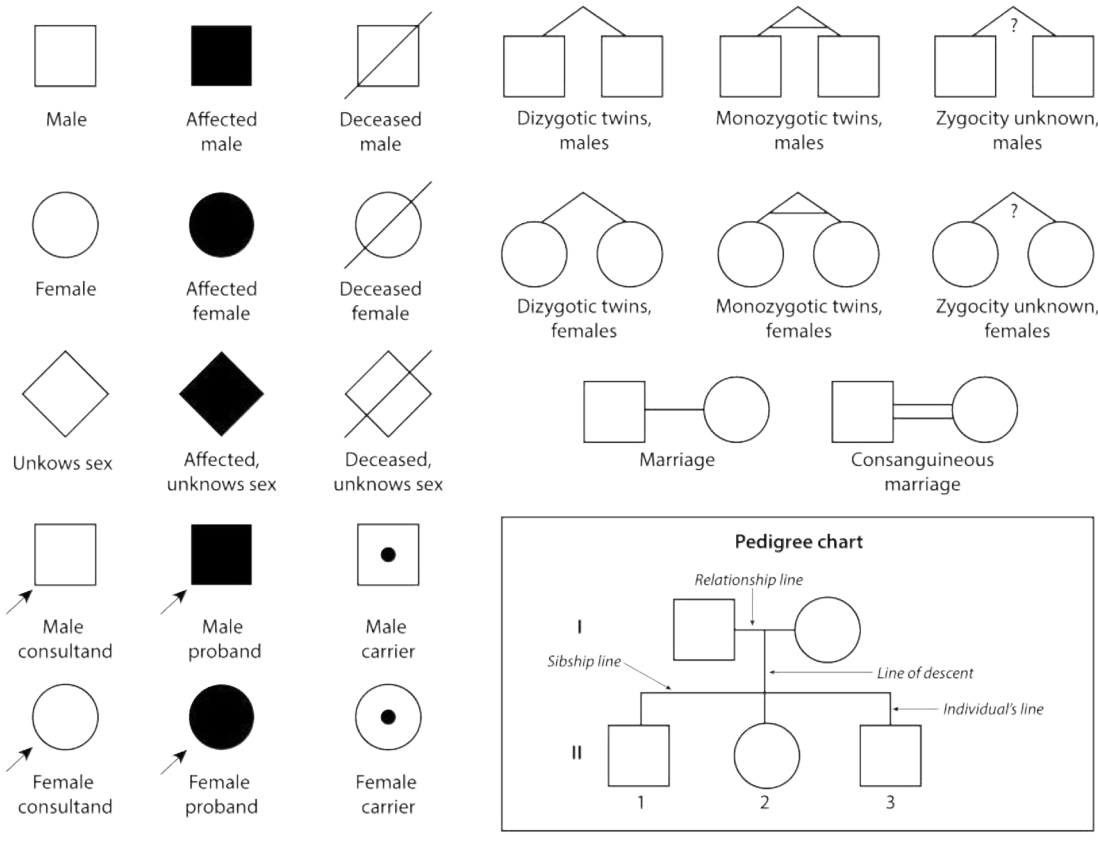
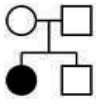


Figure 24.1: Pedigree chart symbols – by international conventions

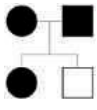
Stick to the conventions when drawing family trees.

INHERITANCE PATTERNS

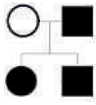
DETERMINING INHERITANCE PATTERNS FROM FAMILY TREE



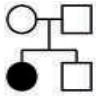
Trait is recessive. Two unaffected parents produce and affected offspring.



Trait is dominant. Two affected parents produce an unaffected offspring.

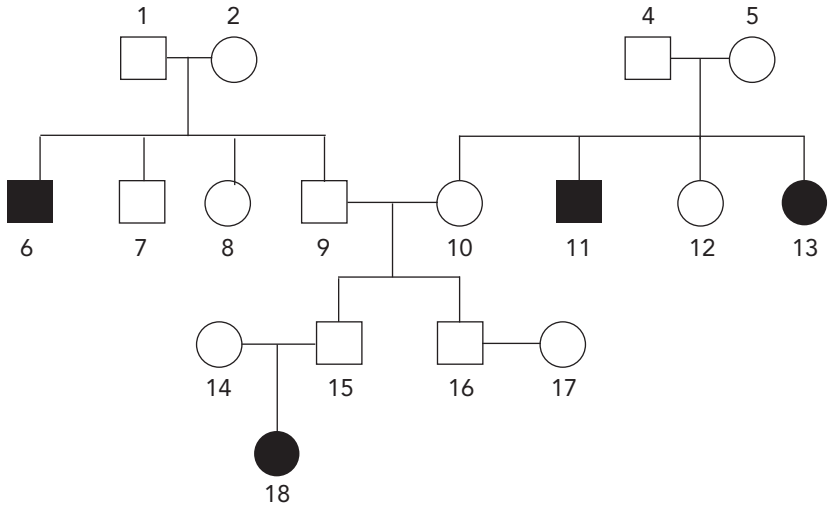


Trait is NOT sex-linked if mother is homozygous.



Trait is NOT sex-linked as father does not have the trait and mother is unaffected.

LOOKING FOR EVIDENCE



- 1 and 2 are unaffected but produce an affected son (6) → recessive trait
- 9 and 10 are unaffected but have a grandchild with the trait (18) → skips a generation so trait is recessive
- 14 and 15 are unaffected but produce an affected daughter (18) → trait is not sex-linked because father is unaffected

MULTI-ALLELIC INHERITANCE CO-DOMINANCE

Blood groups are determined by three alleles. The alleles code for specific glycoproteins found on the surface of red blood cells. The I alleles cause the red blood cells to produce the glycoproteins. The alleles are:

- I^A – red blood cells have glycoprotein A
- I^B – red blood cells have glycoprotein B
- i – red blood cells have no glycoproteins present
- I is dominant over i
- I^A and I^B alleles are co-dominant i.e. cells produce both glycoproteins.

A person can only have two alleles for the trait – one from each parent.

Task 24.1: Write all the possible genotypes for the ABO blood groups that occur in the human population. Remember that a person has only two alleles for their ABO blood group and that the alleles available are I^A, I^B, and i.

Genotype	Phenotype (blood group)

HUNTINGTON'S DISEASE – AUTOSOMAL DOMINANT

- calculating probabilities

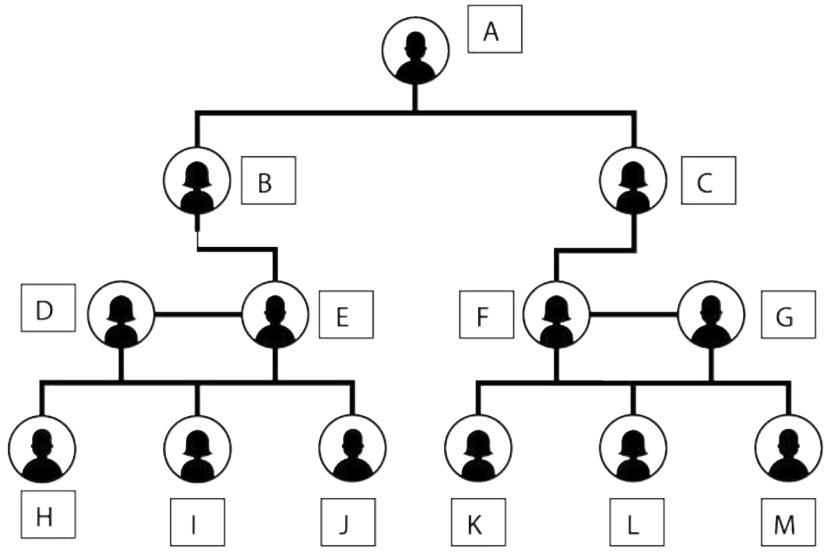


Figure 24.2: Huntington's disease family tree

Person A had Huntington's disease. This family tree shows his descendants.

A has the possibility of being HH or Hh. His mother suffered from Huntington's disease but no-one in his father's family had the disease. Therefore, we can assume his father was hh and his mother could have been HH or Hh.

A has to be Hh – H from his mother and because his father is hh, he must have given h to A.

Assuming A is Hh, the probability of passing on the H allele to his offspring is 0.5.

His daughters, B and C both have 0.5 probability of carrying the H allele.

Each daughter has 0.5 probability of passing the H allele onto each of her children.

Therefore the probability of B and C's children the allele is:

0.5 (prob of B and C having H allele) x 0.5 (prob of father having the H allele) = 0.25

Each child from the same combination of parents has the **same** probability of having the H allele.

Task 24.2: Determine your own phenotype by completing the table below. Does anyone else have the same set of characteristics as you?

Feature and description	Dominance/ recessiveness	Me
Attached ear lobes – ears have no free section below the point of attachment to the head	recessive	
Widow's peak – the hairline forms a distinct point in the centre of the forehead	dominant	
Tongue rolling – ability to roll the tongue into a u-shape tube from front to back	dominant	
Bent little finger – the last joint of the little finger distinctly bends inwards towards the fourth finger when the hands are flat on the table	dominant	
Hitchhiker's thumb – can bend the distal joint of the thumb back to about a 90 degree angle without pressure. It may only be in one thumb.	recessive	
Mid-digital hair – complete absence of hair from the middle phalange of all fingers	recessive	
Index finger shorter than ring finger	Males: dominant Females: recessive	
Interlocking fingers – when fingers are interlocked the left thumb is over the right thumb	dominant	
Cheek dimples	dominant	
Chin dimple/cleft	dominant	
Long eyelashes	dominant	
Wide nostrils	dominant	
Freckles	dominant	

SEX LINKAGE

The X chromosome is larger than the Y chromosome and contains more genes than the Y chromosome. Therefore, some of the genes on the X chromosome have no pair in the Y chromosome. The genes present on the X chromosome are expressed in the phenotype.

For males to have a recessive sex-linked phenotype, they need only one allele for the conditions which is found on their X chromosome.

For females to have the recessive sex-linked phenotype, they need to be homozygous i.e. have two recessive alleles, one on each of their X chromosomes.

Conditions that are sex-linked in humans are

- haemophilia
- colour-blindness
- Fragile X syndrome
- Vitamin D resistant rickets.

The genotypes of sex-linked traits are shown as

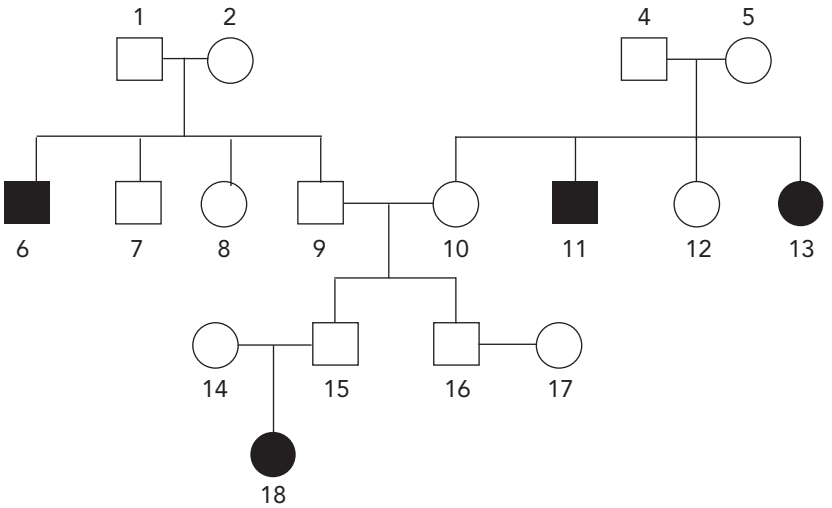
Female $X^A X^A$ – normal Male $X^A Y$ – normal

Female $X^A X^a$ – carrier Male $X^a Y$ – affected

Female $X^a X^a$ – affected

Question 1

Answer the following questions with reference to the following family tree.



(a) Determine the probability of 4 and 5 having two affected offspring. (3 marks)

(b) Calculate the probability of 16 being homozygous dominant. (3 marks)

(c) Determine the probability of 14 and 15 producing an unaffected son as the next offspring. (3 marks)

Question 2

Cystic fibrosis is a genetic disease in which excess mucus accumulates in the lungs and digestive system of affected individuals. Males and females must inherit 2 of the same alleles with this mutation to have the disease. What is the mode of inheritance of cystic fibrosis? Explain your answer. (3 marks)

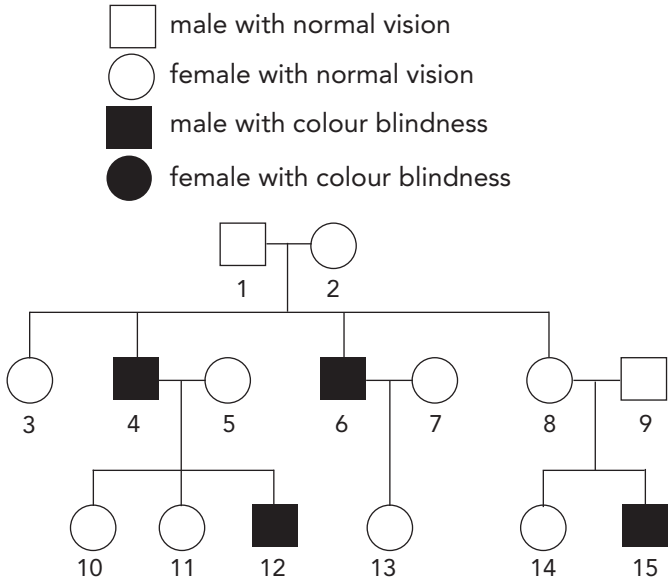
Question 3

Two normal people bred and produced 3 non-albino offspring and 2 albino offspring. Assuming no mutations, who in the family must be heterozygous? Explain your answer. (3 marks)

Question 4

Jeanine inherited 2 alleles for round eye shape and has round eye shape. Her brother inherited 1 allele for round eye shape and 1 allele for almond eye shape and has almond eye shape. Explain the inheritance of eye shape. (4 marks)

Question 5



The pedigree shows 3 generations of a family and the family's incidences of colour blindness, a sex-linked, recessive trait. Which females must have the same genotype? (3 marks)

Question 6

Explain the observation that females rarely get the disease haemophilia. (2 marks)

Question 7

What could be the blood type of a person whose parents were A and O blood types? (3 marks)

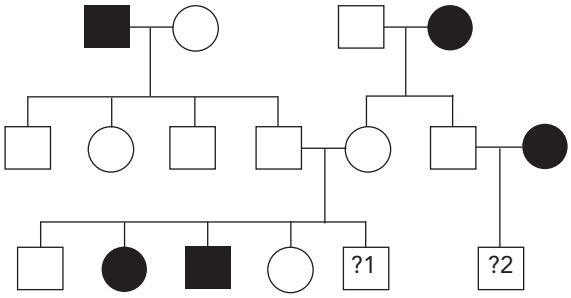
Question 8

While studying several generations of a particular family, a geneticist observed that a certain disease was found equally in males and females and that all children who had the disease had parents who also had the disease. Explain the gene coding for this disease. (2 marks)

Question 9

If both parents carry the recessive allele that causes cystic fibrosis, calculate the probability of their child will develop the disease. (3 marks)

Question 10

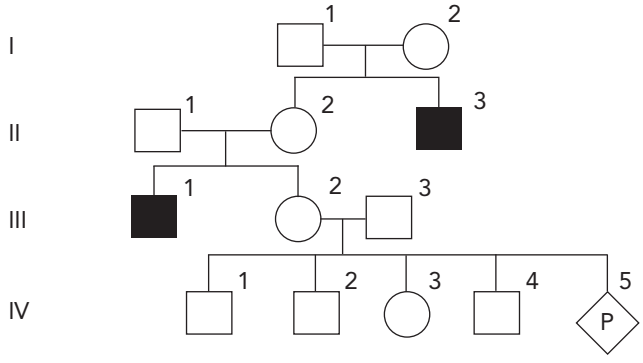


(a) Calculate the probability of ?1 carrying the affected allele. (3 marks)

(b) If the child marked ?2 was NOT affected by the condition. What would you tell the father of that child about his and his son's genotype? (2 marks)

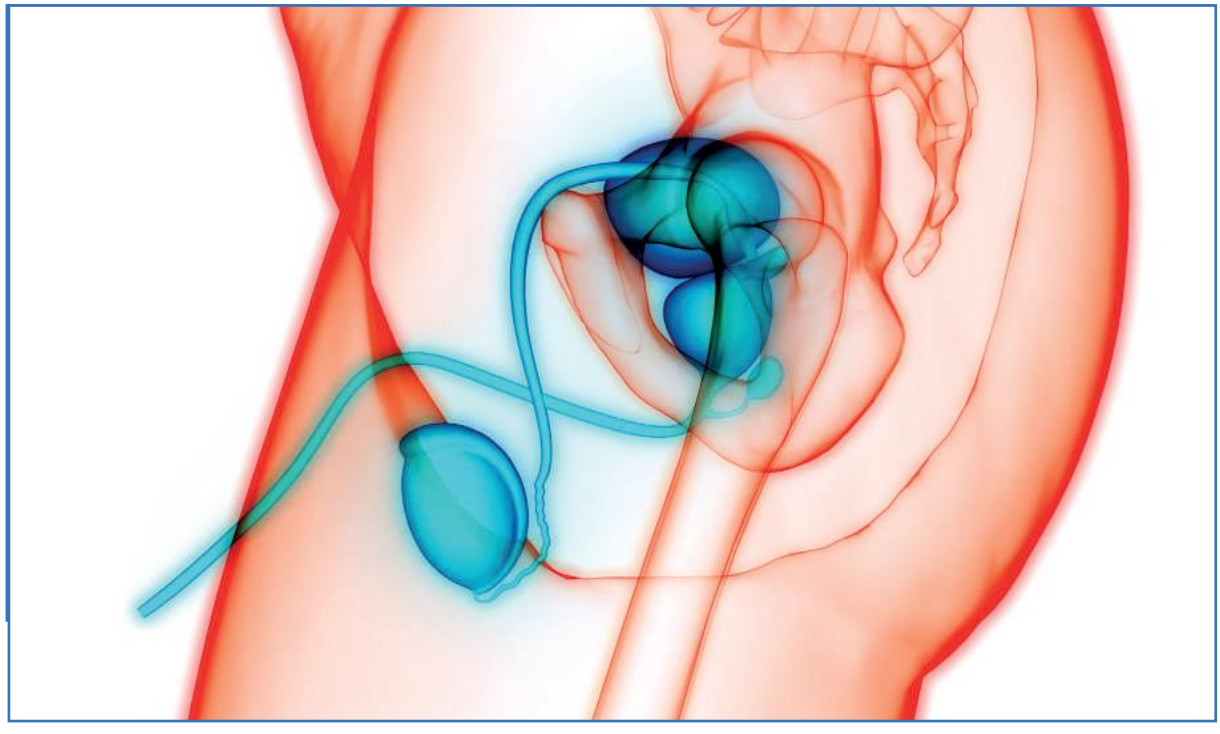
Question 11

The following is a family tree showing the incidence of an X-linked condition. (3 marks)



Show how to calculate the probability of P being affected.

25 MALE REPRODUCTIVE SYSTEM

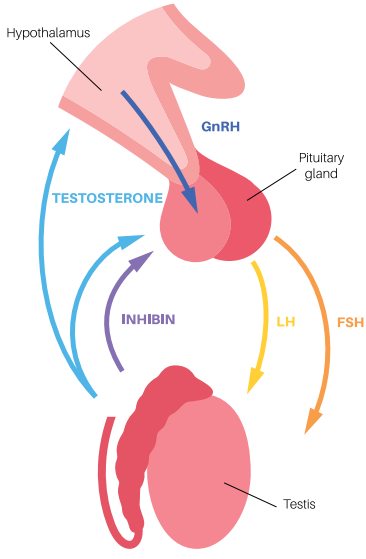


Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The production of offspring is facilitated by the structure and function of the male and female reproductive systems in producing and delivering gametes for fertilisation and providing for the developing embryo and foetus.				
• Both male and female reproductive systems are regulated by hormones, including the regulation of the menstrual and ovarian cycles.				

The male reproductive systems has three functions:

- production of male hormones
- production of male gametes
- sperm delivery system.

PRODUCTION OF MALE HORMONES



GnRH – gonadotrophin

FSH – follicles stimulating hormone

LH – luteinising hormone

BUT males don't have follicles!

No, they don't but **FSH** stimulates testicular growth and sperm production in males.

GnRH – promotes the synthesis and secretion of FSH and LH.

LH – promotes the production of testosterone from Leydig or interstitial cells in the testes.

Figure 25.1: Male reproductive hormones

Testosterone is a sex hormone and anabolic steroid that is produced mainly in the testicles of males. It regulates:

- sex drive
- bone mass
- fat distribution
- muscle mass and strength
- the production of red blood cells and sperm.

Testosterone binds to and activates the androgen receptor, which influences the development of male reproductive tissues and secondary sexual characteristics, such as the penis, testes, voice, body hair, and muscle and bone mass.

Task 25.1: Link the development of the secondary sex characteristics to the functions in the body that are regulated by testosterone.

Testosterone also affects health and well-being, including moods, behaviour, and the prevention of osteoporosis.

Testosterone levels usually decline with age.

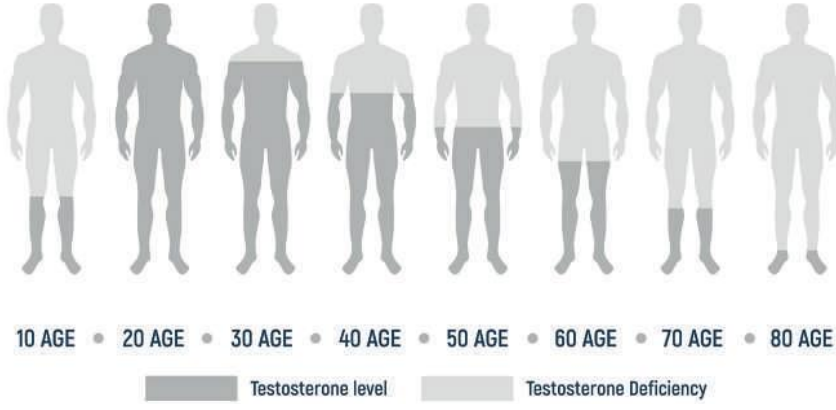


Figure 25.2: Testosterone levels in males at different ages

PRODUCTION OF MALE GAMETES

Male gamete (sperm) formation occurs in the seminiferous tubules which are extremely long, which:

- allows time for the spermatozoa to mature before leaving the testes.
- provides the surface area for the number of parent cells required to produce the millions of gametes in the semen.

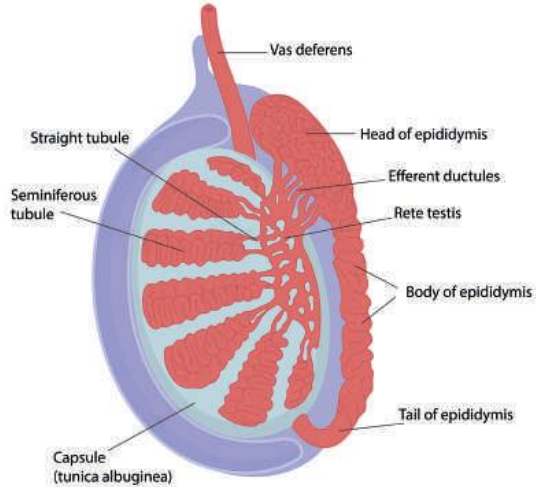


Figure 25.3: Testicular structure

The male gametes are stored in the epididymis until they are required during ejaculation. While in the epididymis, they develop their final form ready to fertilise an ovum should the opportunity arise.

The parent cells lining the seminiferous tubules undergo meiosis. At each stage the cells move towards the centre of the tubules from where they can move to the epididymis for storage.

The sequence is shown below in Figure 25.4.

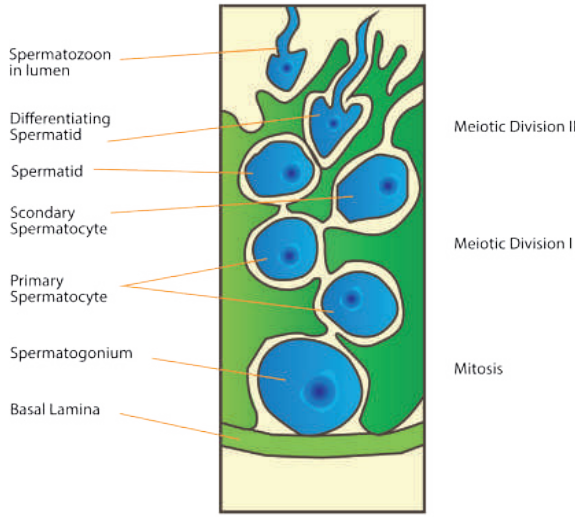


Figure 25.4: Development of sperm in the lining of the seminiferous tubule

The process of spermatogenesis takes about 74 days. Sperm have a life span of about 2–3 months in the epididymis, then, if there is no ejaculation, they degenerate and are reabsorbed into the body.

Testes produce 200 to 300 million spermatozoa daily. However, only about half or 100 million of these become viable sperm.

The process of spermatogenesis in not perfect and the resultant sperm maybe of different forms as shown in Figure 25.5.

The testes are located outside the male body because the process of spermatogenesis and sperm maturation require a lower temperature than normal body temperature.

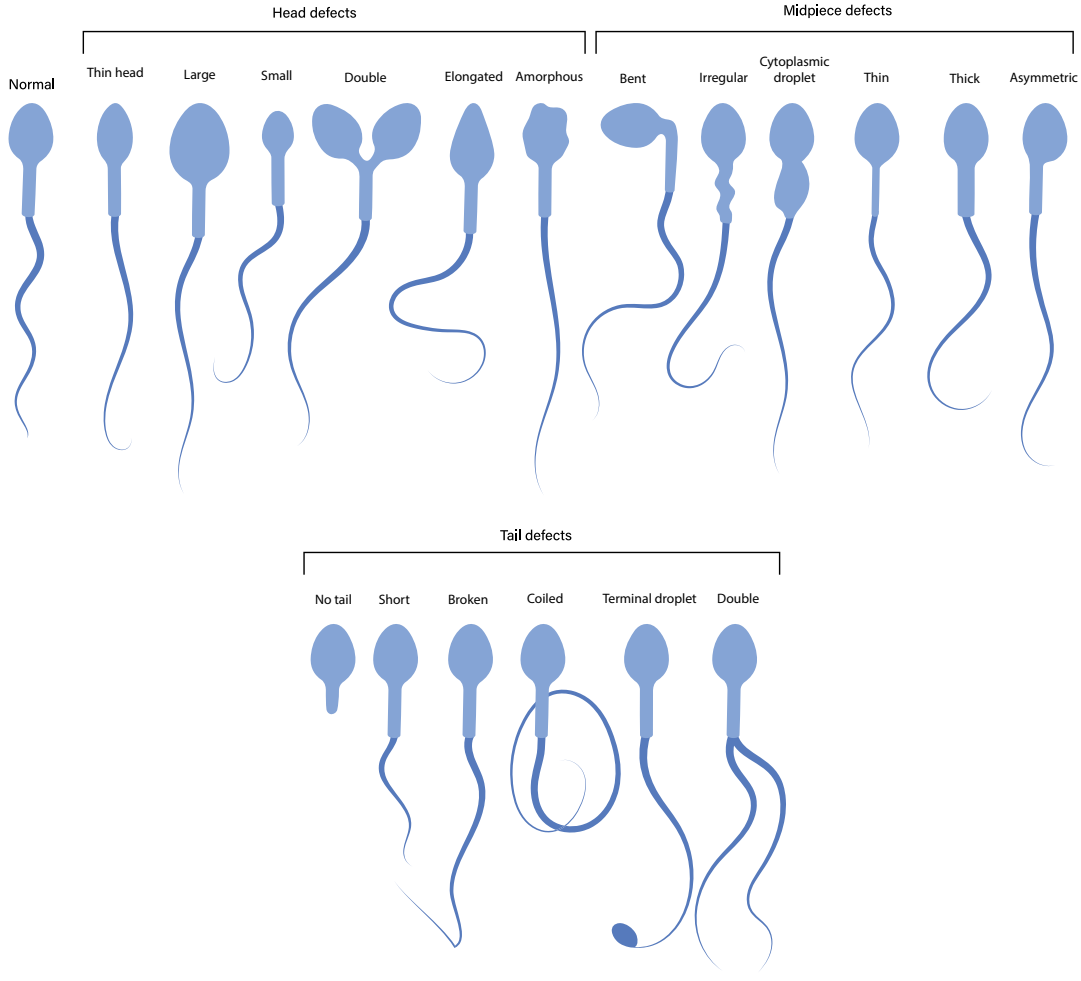


Figure 25.5: Human sperm morphologies

Task 25.2: Explain how the differences could impact on the viability of the sperm.

SPERM DELIVERY SYSTEM

The male reproductive system is a series of tubules.

Different glands add materials to the tubules at different times.

- The fluid from the seminal vesicle contains fructose for an energy source, protein for coagulation reaction and prostaglandins for mobility and viability of the sperm.
- Prostate gland secretes alkaline fluid. Having the optimum pH level in semen is important to keep sperm cells healthy and mobile, while the alkalinity helps to neutralise the acidic environment of the vagina.

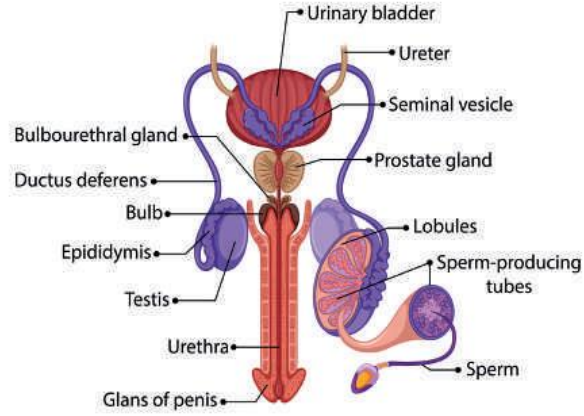


Figure 25.6: Cross section of the male reproductive system

The urethra is used by the:

- male reproductive system to carry semen from the vas deferens to the outside
- urinary/excretory system to carry urine from the bladder to the outside

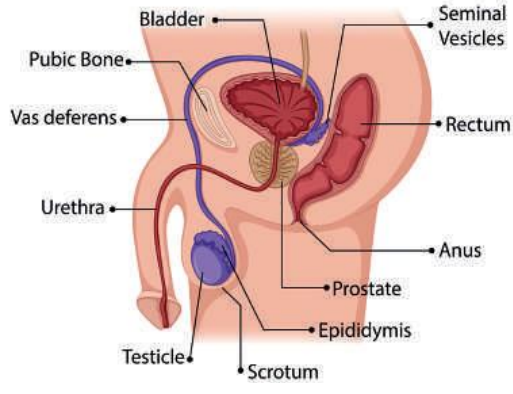


Figure 25.7: Sagittal section of the male reproductive system

For fertilisation to occur, sperm need to travel from the epididymis in the testes in the male to the upper end of the vagina in the female.

The sperm need to arrive in a condition that allows them the swim against the fluid currents in the uterus and fallopian tubes to the ovum and then to be able to penetrate the outer layer of the corona radiata to get to the ovum itself.

An erection occurs when the penis becomes hard and enlarged from an increase in blood flow.

Two chambers called the corpora cavernosa, run the length of the penis and contain a maze of blood vessels shaped like a sponge. The sponge-like structure, allows blood to fill the open space producing an erection. The penis in this condition is able to penetrate the vagina ready for ejaculation.

Ejaculation is when semen is ejected from the penis. It has two phases:

1. Emission phase – sperm move from the testes to the prostate area where fluids from the prostate gland and seminal vesicles are added to produce semen. The vas deferens contracts to move the semen to the base of the penis.
2. Expulsion phase – the muscles at the base of the penis contract, expelling the semen out of the urethra.

Question 1

Where are sperm produced? (1 mark)

(a) Prostate gland
(b) Epididymis
(c) Vas deferens
(d) Seminiferous tubules

Question 2

Which chemical or hormone produced by the testes causes bodily changes during male puberty. (1 mark)

(a) FSH
(b) LH
(c) Testosterone
(d) Gonadotropin

Question 3

The testes are situated outside the abdominal cavity within a pouch called the scrotum. This is necessary because: (1 mark)

(a) The scrotum contains very long ducts and tubules for the transfer of sperm.
(b) The external location helps to maintain the lower temperature required for spermatogenesis.
(c) The scrotum reduces the pressure around the testes to aid in the rate of spermatogenesis.
(d) Large amounts of sperm and semen are stored in the flexible scrotal sac.

Question 4

Which of the following male organs is considered the primary sex organ? (1 mark)

(a) Penis
(b) Testes
(c) Prostate gland
(d) Epididymis

Question 5

The testes secrete testosterone from which of the following? (1 mark)

(a) Seminiferous tubules
(b) Prostate gland
(c) Epididymis
(d) Interstitial cells

Question 6

Which of these is not a function of male sex hormones? (1 mark)

(a) Deposition of fats on the chest and thighs
(b) Growth of bone and muscle
(c) Growth of facial and body hair
(d) Development of male external genitalia

Question 7

Which organ releases gonadotrophin in males? (1 mark)

(a) Anterior pituitary
(b) Posterior pituitary
(c) Hypothalamus
(d) Testes

Question 8

What does follicle-stimulating hormone stimulate in males? (1 mark)

(a) Production of sperm
(b) Release of testosterone
(c) Release of gonadotropin-releasing hormone
(d) Release of luteinising hormone

Question 9

Name the site of sperm maturation? (1 mark)

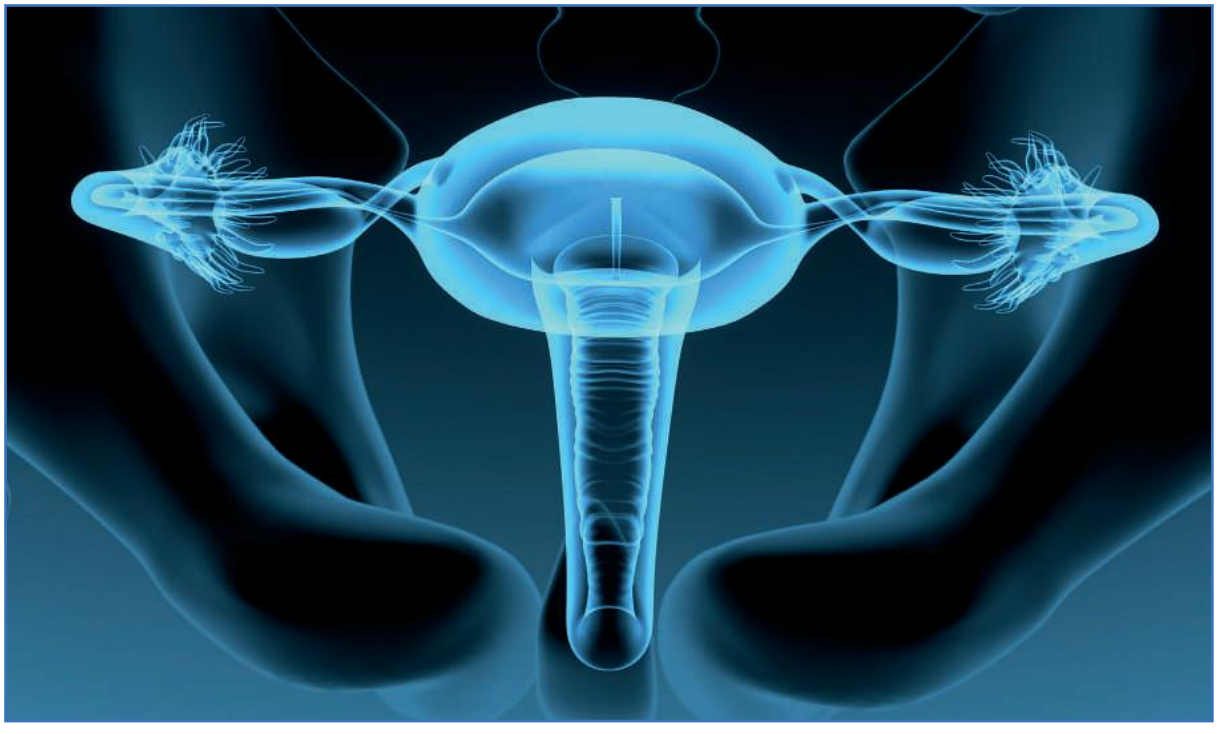
(a) Epididymis
(b) Ductus deferens
(c) Spermatic cord
(d) Urethra

Question 10

Outline the functions of the chemical contents of the semen. (4 marks)

FEMALE REPRODUCTIVE SYSTEM

26



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The production of offspring is facilitated by the structure and function of the male and female reproductive systems in producing and delivering gametes for fertilisation and providing for the developing embryo and foetus.				
• Both male and female reproductive systems are regulated by hormones, including the regulation of the menstrual and ovarian cycles.				

The female reproductive systems has three functions:

- production of female hormones
- production of ova
- support and nutrition of developing embryo/foetus

The structure of the female reproductive system provides for:

- the production of female gametes (ovary)
- a pathway for sperm to travel to the ovum (vagina, uterus and fallopian tube)

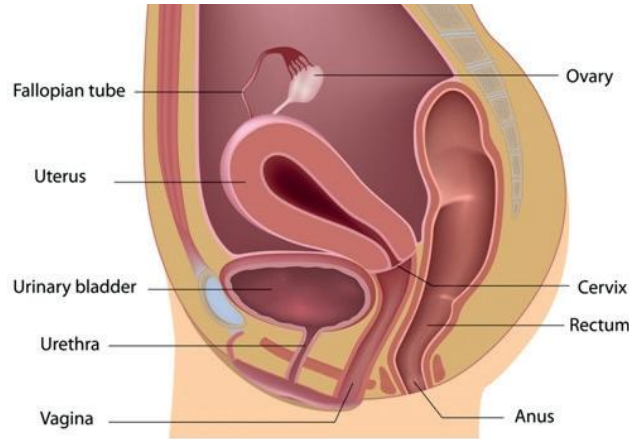


Figure 26.1: Sagittal section of the female reproductive system

- a pathway for the ovum to move from the ovary to the uterus (fallopian tube)
- a place for the development and support of the growing embryo/foetus (uterus, cervix)
- a pathway for the foetus to exit the uterus (vagina).

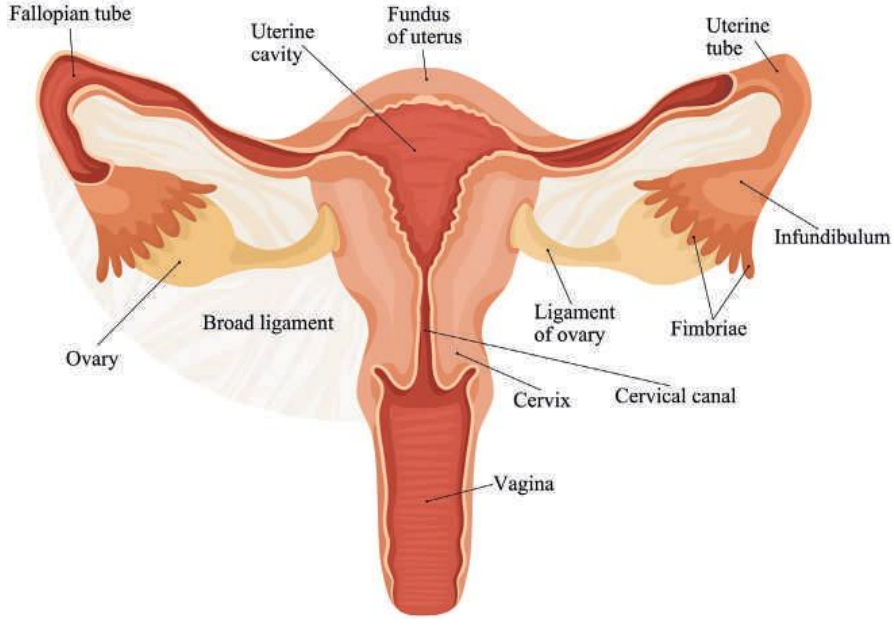


Figure 26.2: Coronal section of the female reproductive system

The **fimbriae** end of the fallopian tube moves across the ovary to cover the location of the developing follicle to 'catch' the ovum as it erupts from the follicle during ovulation.

The **fallopian** tubes provide a pathway for the ovum to the uterus and a place for fertilisation supported by secretions that form fluid currents to help the movement of the ovum. They are lined with cilia which keep the current moving towards the uterus.

The **cervix** is a strong muscular sphincter that provides support for the developing embryo and reduces the chances of miscarriage.

The **uterus** is a strong muscular structure that is capable of huge expansion as the foetus grows; it contracts strongly during labour and childbirth.

Ligaments surrounding the structures keep them in place within the abdominal cavity.

OVARIAN CYCLE

The ovarian cycle involves the changes that occur in the ovary, bringing about the maturation of an ovum for ovulation and, possibly, fertilisation.

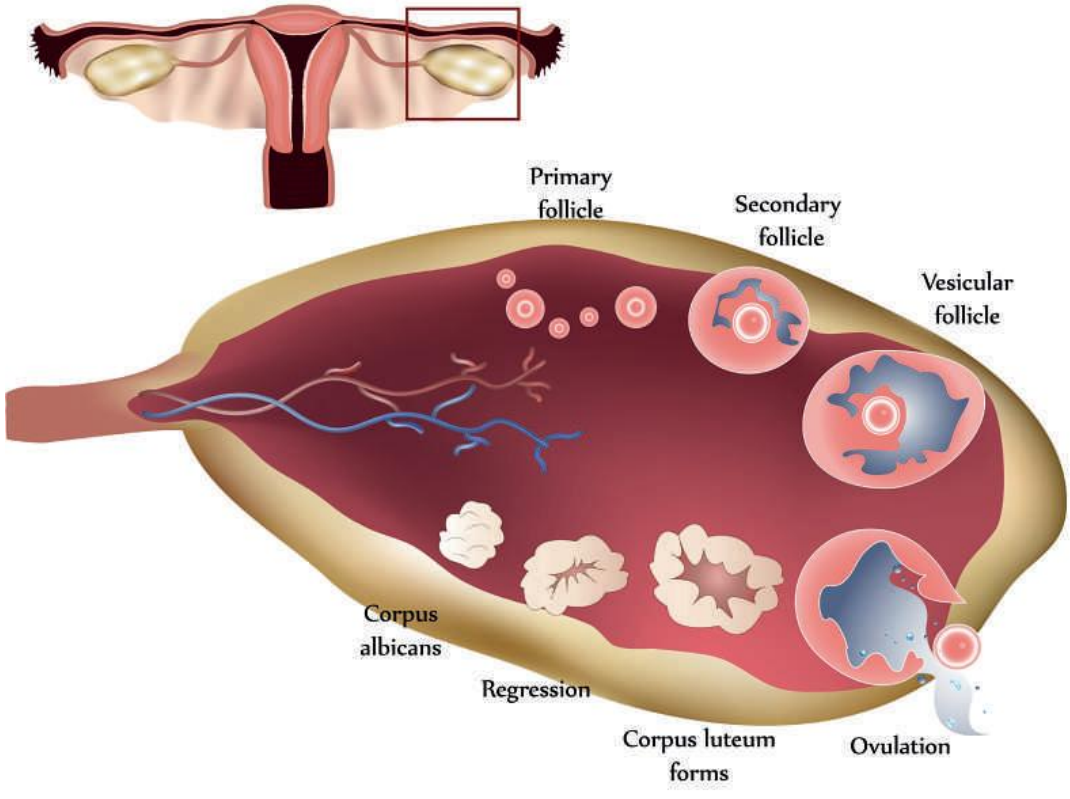


Figure 26.3: Ovarian cycle

Primary follicle – contains cell at the early stages of meiosis; remains dormant until influenced by female hormones; has a single layer of follicle cells.

Secondary follicle – cell undergoing the final stages of meiosis, may have follicular fluid and has many layers of follicle cells.

Graafian/vesicular follicle – contains the mature ovum and layers of follicle cells and follicle fluids. It is located near the surface of the ovary ready for ovulation.

Corpus luteum – growth of cells in the space left by the release of the ovum from the Graafian follicle. It produces progesterone.

Corpus albicans – is the degenerating corpus luteum and may leave a scar on the surface of the ovary.

Ovulation – the rupture of the Graafian follicle to release the ovum and follicle fluids into the fallopian tube.

Figure 26.3 shows all the stages of the ovarian cycle in the ovary, but this doesn't happen all at the same time. Only the primary follicles and one other stage occurs at one time in an ovary.

THE OVUM

The **ovum** is a large, immobile, spherical cell that consists of the nucleus, the cytoplasm and the cellular membrane The nucleus contains one-half of the genetic materials (a haploid number of chromosomes), and the cytoplasm stores the nutrients for the early embryonic development.

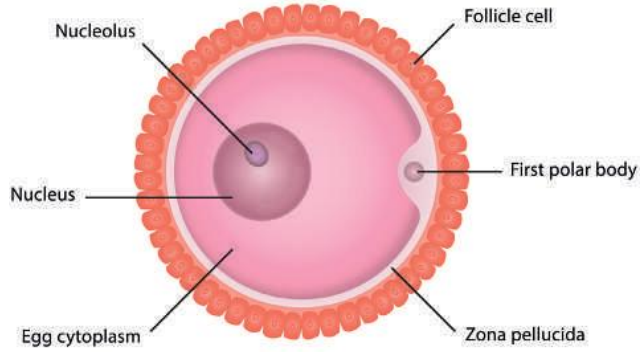


Figure 26.4: Structure of the ovum

A **polar body** may be present as a result of meiosis, but it usually degenerates and is reabsorbed.

The **zona pellucida** is a specialised extracellular matrix that surrounds the plasma membrane of oocytes. It is a vital constitutive part of the ovum in preventing polyspermy – entry of more than one sperm – and enables the acrosome enzyme reaction for the successful adhesion and penetration by one sperm cell. It also allows for the correct genetic content. If the zona pellucida ruptures in later development stages, the embryonic cells could separate, forming multiple embryos.

The **corona radiata** is composed of follicle cells that provide protection for the ovum when it is ejected from the ovary.

OVARIAN CYCLE

The ovarian cycle involves the changes in the ovarian follicle in which the developing ovum forms.

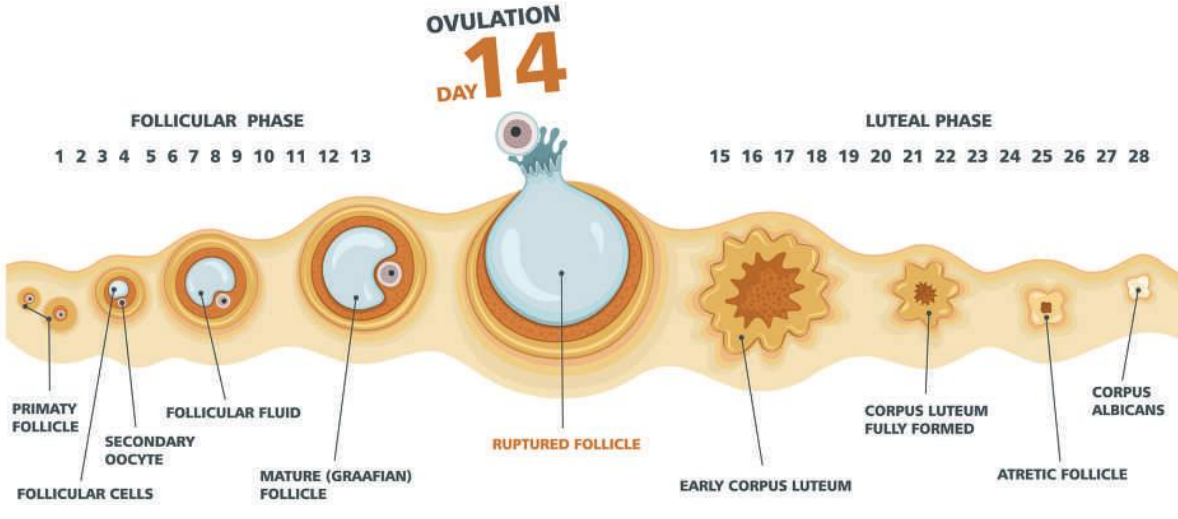


Figure 26.5: Timing of ovarian cycle

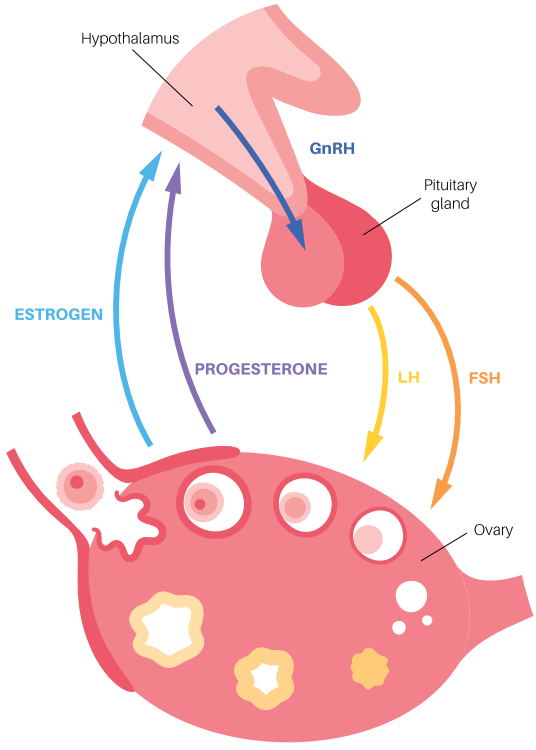


Figure 26.6: Pituitary and ovarian hormones

The ovarian cycle is controlled by hormones from the hypothalamus and anterior pituitary.

HYPOTHALAMUS

- **GnRH** – gonadotrophic releasing hormone – promotes the production of FSH and LH in the anterior pituitary.

ANTERIOR PITUITARY

- **FSH** – follicle stimulating hormone – regulates the development, growth, pubertal maturation, and reproductive processes of the body; specifically stimulates the development of the ovarian follicles.
- **LH** – luteinizing hormone – promotes the final maturation of the ovarian follicle, ovulation and formation of the corpus luteum.

OVARIAN HORMONES

Oestrogen – produced by the ovarian follicle during development, reaching a maximum level prior to ovulation. With the sharp fall of oestrogen concentration, FSH and LH production increases greatly, promoting ovulation. After ovulation the corpus luteum produces oestrogen inhibiting FSH and LH production. Oestrogen is also responsible for the development of the female secondary sex characteristics, such as:

- development and growth of breasts
- growth of body hair, most prominently underarm and pubic hair
- widening of hips, resulting in a lower waist to hip ratio than adult males
- deposition of fat, predominantly in the breasts, hips, and thighs
- broadening of the pelvis and growth of the uterus and vagina.

Progesterone – produced by the corpus luteum, it inhibits FSH and LH production so no more follicles will develop during this time. Progesterone promotes the thickening of the endometrium so it will be receptive to a fertilised ovum.

A decrease in the concentrations of oestrogen and progesterone is caused by the breakdown of the corpus luteum and, by negative feedback, increases the production of FSH and LH to start the next cycle.

MENSTRUAL CYCLE

The **menstrual cycle** is a monthly process that prepares the uterus for pregnancy.

The four phases of the menstrual cycle are:

- **Menstrual phase** – starts when an egg from the previous cycle isn't fertilised. Levels of oestrogen and progesterone drop, and the thickened lining of the uterus sheds through the vagina. It aligns with the breakdown of the corpus luteum in the ovarian follicle.
- **Follicular phase** or proliferative stage phase – the thickening of the uterine lining. It aligns with the production of the Graafian follicle containing the mature ovum.
- **Ovulation phase** – the time of maximum development of the uterine lining. It aligns with the releases a mature ovum from the Graafian follicle (ovulation) which travels down the fallopian tube toward the uterus to be fertilised.
- **Luteal** or secretory phase – the time between ovulation and the start of the next period when the uterine lining is maintained ready for the implantation of a fertilised ovum. It aligns with the production of progesterone from the corpus luteum.

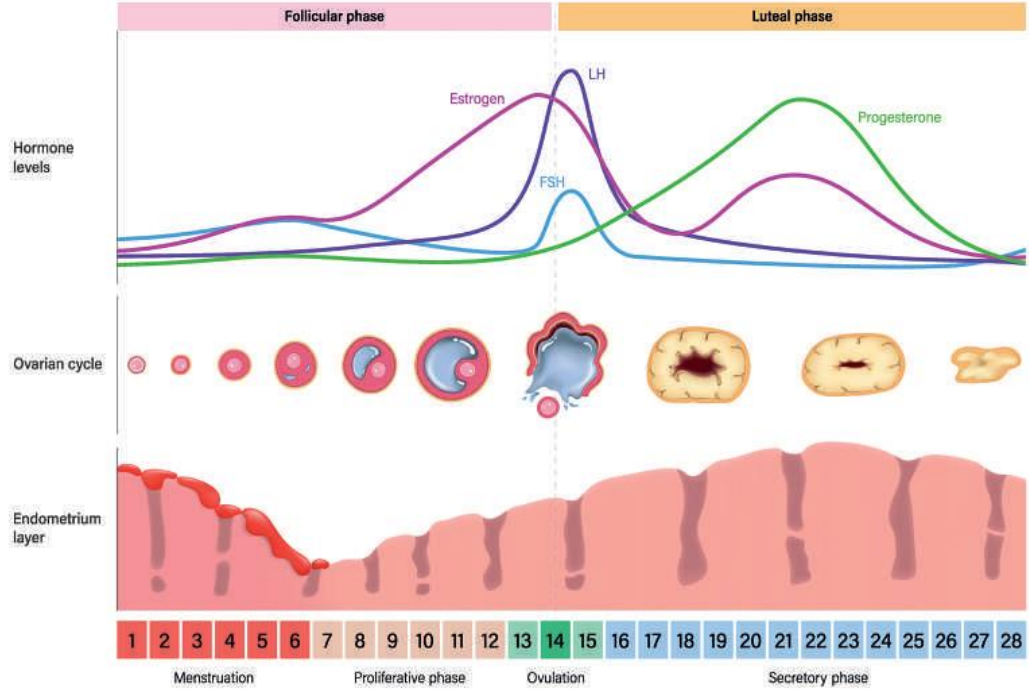


Figure 26.7: Changes during the menstrual cycle

Question 1

Explain why are the ovaries called the primary sex organ of the female reproductive system. (1 mark)

Question 2

Describe the functions of the fallopian tubes. (2 marks)

Question 3

Describe the feedback mechanism that controls the production of FSH. (5 marks)

Question 4

Explain why it is important that the zona pellucida prevents polyspermy. (2 marks)

Question 5

Describe what happens to the ovary and the uterus if the ovum is not fertilised. (4 marks)

Question 6

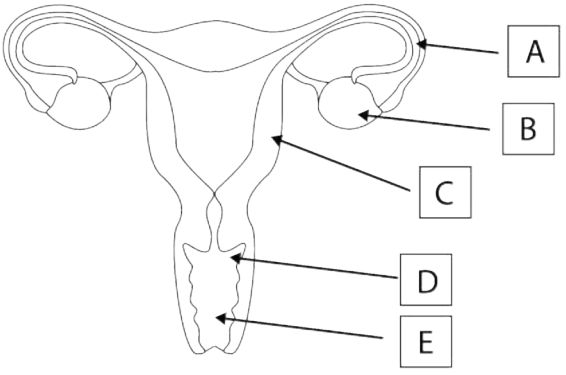
The ovum ejected from the ovarian follicle at ovulation cannot move independently. Describe how it moves through the fallopian tube to the uterus. (2 marks)

Question 7

Describe contents of the menstrual flow. (1 mark)

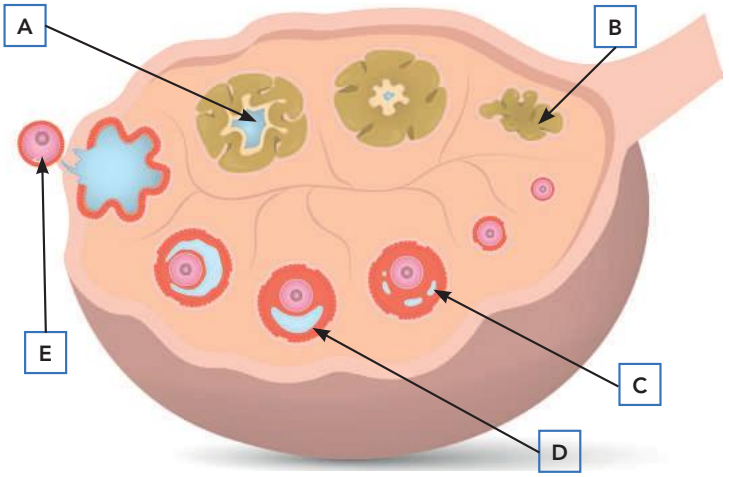
Question 8

Label the following diagram. (5 marks)



Question 9

Label the following diagram showing the changes during the ovarian cycle. (5 marks)



CONTRACEPTION

27

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Contraception methods that reduce the probability of the union of gametes or implantation all have limitations, risks and benefits, and include methods that:				
o use fertility awareness				
o use steroid hormones				
o use physical barriers between gametes				
o use chemical spermicides				
o use sterilisation (tubal ligation, vasectomy)				
o function after coitus (emergency contraceptive pill and intrauterine devices [IUDs]).				

Contraception is the prevention of pregnancy.

There are many different ways to prevent pregnancy and all have varying effectiveness.

PREVENTING FERTILISATION BY PHYSICAL MEANS

- Abstinence – no sex so sperm is not in the same location as the ovum
- Coitus interruptus – withdrawal of penis before ejaculation
- Rhythm method – no sex during the time the ovum is viable i.e. around ovulation
- Condoms – male or female – prevent the sperm from entering the female reproductive tract
- Diaphragm – usually used with spermicide to block the passage of sperm through the cervix

PREVENTING FERTILISATION BY CHEMICAL (HORMONAL) MEANS

These usually involve females taking hormones to control the maturation and ovulation of the ovum.

- Includes oral contraception – hormone pill taken each day
- Contraceptive injection – longer lasting hormone dose
- Hormonal implant or ring and contraceptive skin patch – continuous release of hormones to control the ovarian cycle

PREVENTING IMPLANTATION

Intrauterine device (IUD) with hormones – placed in the uterus where it thickens mucus in the cervix to stop sperm from reaching or fertilising the ovum, thins the lining of the uterus to reduce chances of implantation and partially suppresses ovulation.

SURGICAL CONTRACEPTION (STERILISATION)

- Females – the fallopian tubes are cut and tied off to stop the sperm from reaching the ovum and ovum from reaching the uterus.
- Males – vas deferens is cut and tied to stop sperm from being delivered to the urethra and becoming part of the semen.

OTHER METHODS

- Douche – is the flushing of the vagina after sexual intercourse to remove semen/sperm.
- 'Morning after pill' – stops or delays ovulation; does not end a pregnancy that has implanted.
- RU-486 pill – works to terminate early pregnancy by forcing the uterus to 'expel' the pregnancy in a way that mimics a woman's regular menstrual period.
- Termination/abortion – the most common type of abortion is a surgical procedure called a 'suction curette'. This involves removing the lining and the contents of the uterus by applying gentle suction to the inside of the uterus.

Task 27.1: Use the information above and the in Figures below to compare the effectiveness of different types of contraception.

Task 27.2: List the factors that could impact on the effectiveness of different methods of contraception.

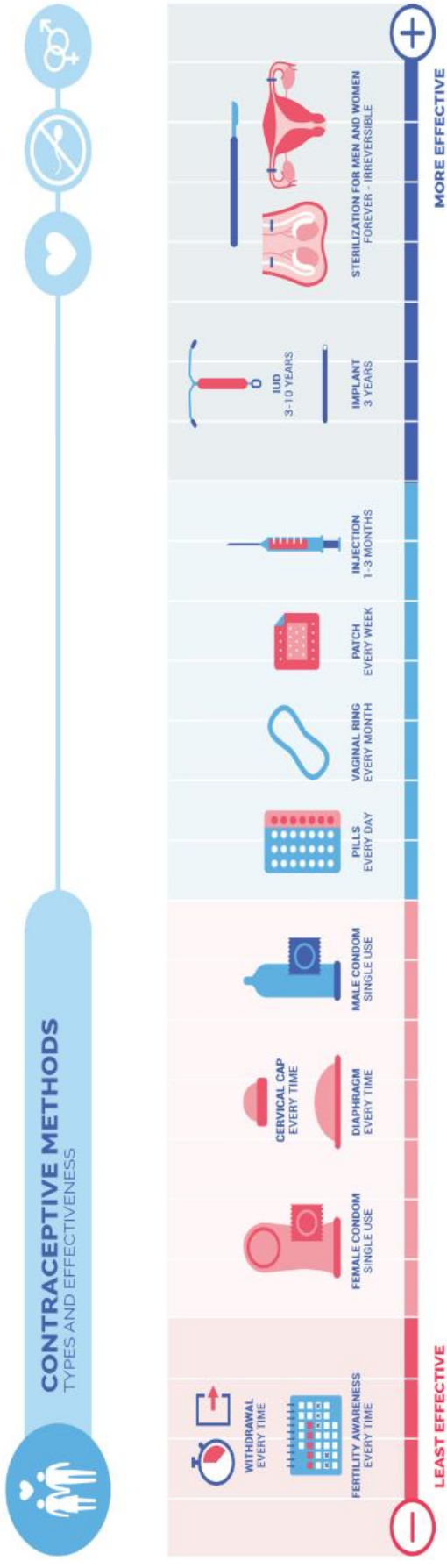


Figure27.1: Contraceptive methods ranked according to effectiveness

Figure 27.2: Contraceptive methods indicating effectiveness, cost and usage

SEXUALLY TRANSMITTED INFECTIONS

28

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Sexually transmitted infections (STIs), diseases transmitted through unprotected sex or genital contact, can be prevented through safe sex methods. Early detection and treatment of infection are important and, if left untreated, STIs can lead to serious health consequences.				

STI cases should be reported in accordance with **state and local statutory requirements**. Syphilis (including congenital syphilis), gonorrhoea, chlamydia, chancroid, and HIV are reportable diseases in every state of Australia.

A **sexually transmissible infection** (STI) is an infection that can be passed from one person to another, usually by unprotected sexual contact, including vaginal, anal and/or oral sex.

Table 28.1: STIs caused by different pathogens

Bacterial STIs	Viral STIs	Fungal STIs	Parasitic STIs
Chlamydia	Herpes	Candidiasis	Trichomonas vaginalis
Gonorrhoea	Genital warts		Pubic lice
Syphilis	HIV		Scabies
Bacterial vaginosis	Hepatitis		

In Western Australia, chlamydia and gonorrhoea are the most common STIs, particularly among teenagers and young adults. Western Australia is experiencing a syphilis outbreak. From 2014 to September 2022, the annual number of infectious syphilis notifications increased nine-fold. (Communicable Disease Control Directorate Alert to physicians 4 January 2023)

For every person diagnosed with an STI, there is at least one other infected person. The more sexual partners a person has, the greater the risk of catching one or more STIs, or of passing on the infection to others.

No immunity is developed to bacterial STIs, so a person can catch them again and again, even after treatment.

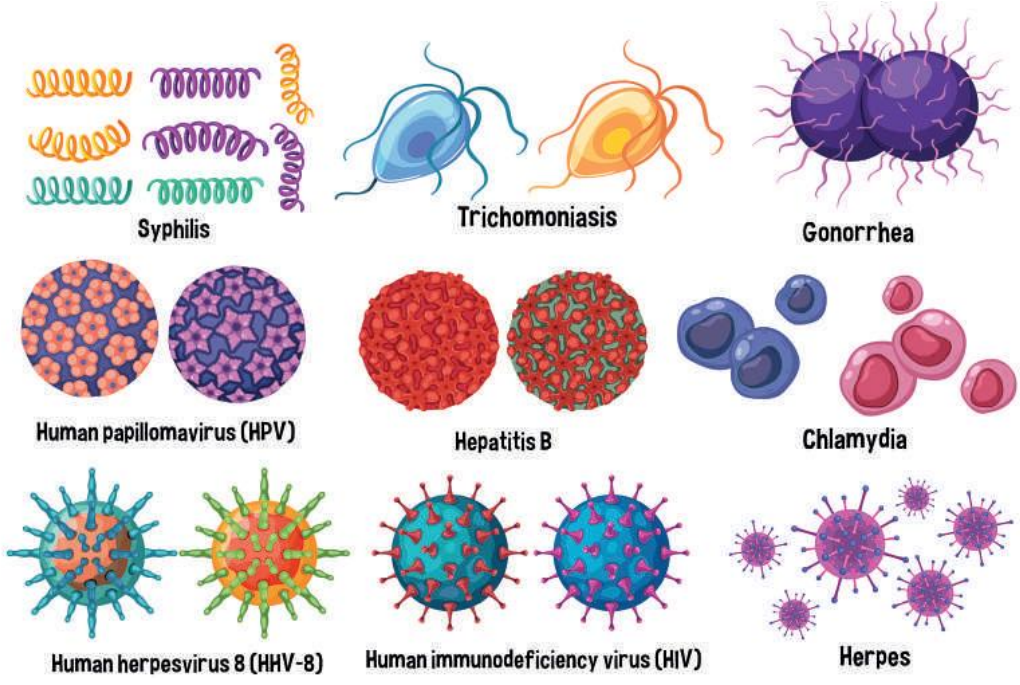


Figure 28.1: Pathogens causing STIs

HERPES

There are two types of the Herpes Simplex Virus (HSV). HSV-1 -usually causes blisters around the mouth and lips, commonly known as cold sores. HSV-2 usually causes blisters around the genitals and rectum.

There is no known cure, but it can be treated.

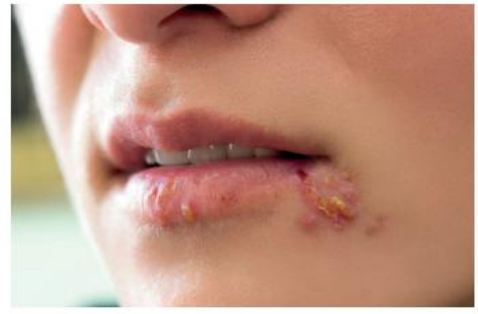


Figure 28.2: Facial herpes HSV-1

SYPHILIS

Syphilis is a bacterial infection. The disease starts as a painless sore, typically on the genitals, rectum or mouth. It is treated with antibiotics.

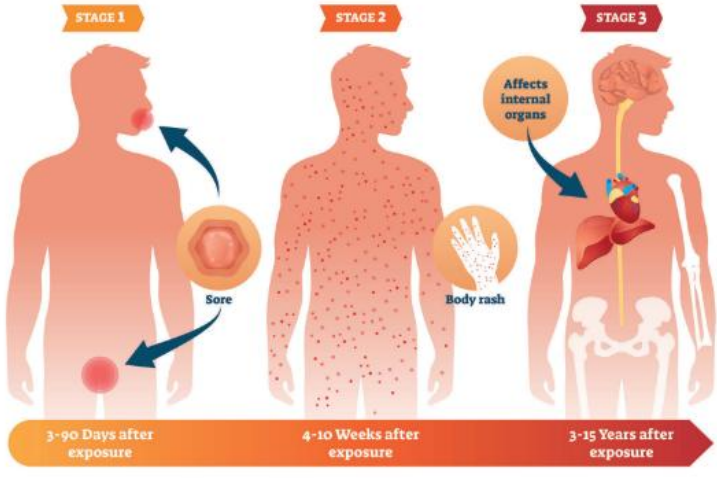


Figure 28.3: Stages of syphilis

A mother can pass syphilis to her baby during the pregnancy or at the time of birth. Congenital syphilis can cause miscarriage or stillbirth. It can also cause other serious health problems in babies including organ, brain or nerve damage.

GONORRHOEA

Often called 'the clap', gonorrhoea is caused by a bacterium.

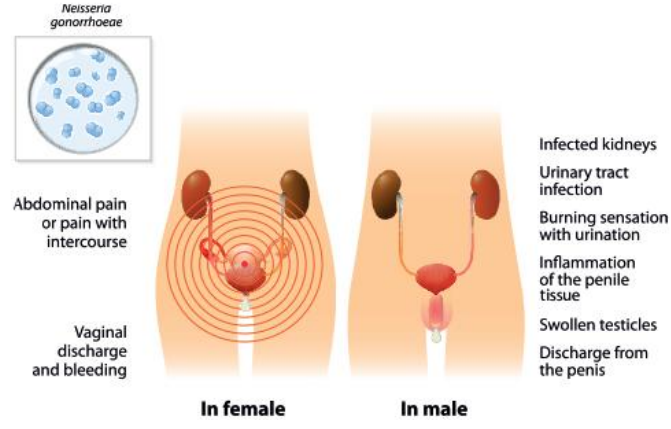


Figure 28.4: Symptoms of gonorrhoea

Treatments by anti-bacterial preparations are available but there is a high level of resistance in the common bacterial strains.

CHLAMYDIA

This infection caused by bacteria. There are no symptoms in the initial stages, but can cause health problems at a later stage, including infertility in females.

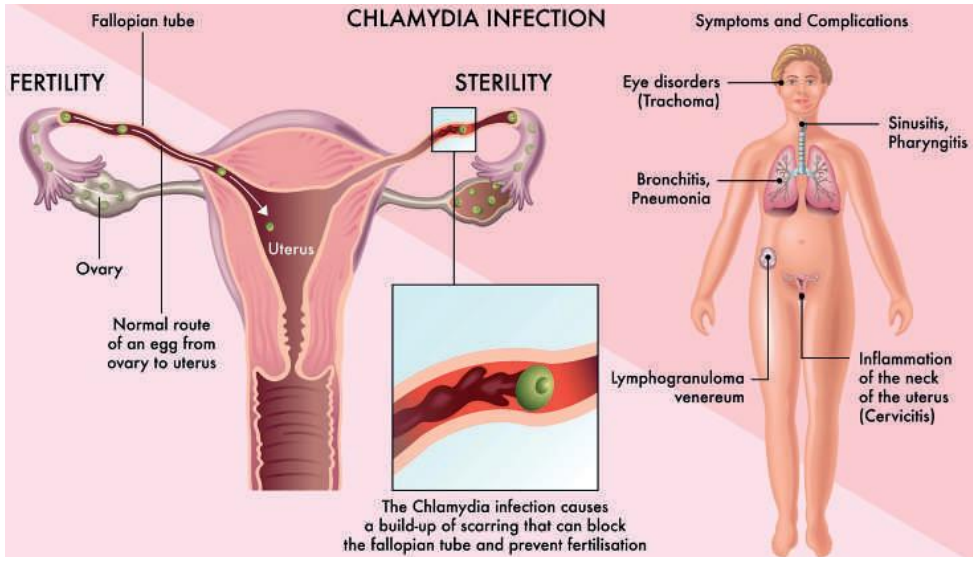


Figure 28.5: Chlamydia infection

Once diagnosed, chlamydia is easy to treat with antibiotics.

HIV AIDS

- **HIV** (human immunodeficiency virus) causes **AIDS** (acquired immunodeficiency syndrome)
- HIV is an infection that attacks the body's immune system. AIDS is the most advanced stage of the disease.
- HIV targets the body's white blood cells, weakening the immune system. This makes it easier to get sick with diseases like tuberculosis, infections and some cancers.
- HIV is spread from the body fluids of an infected person, including blood, breast milk, semen and vaginal fluids. It can also spread from a mother to her baby.

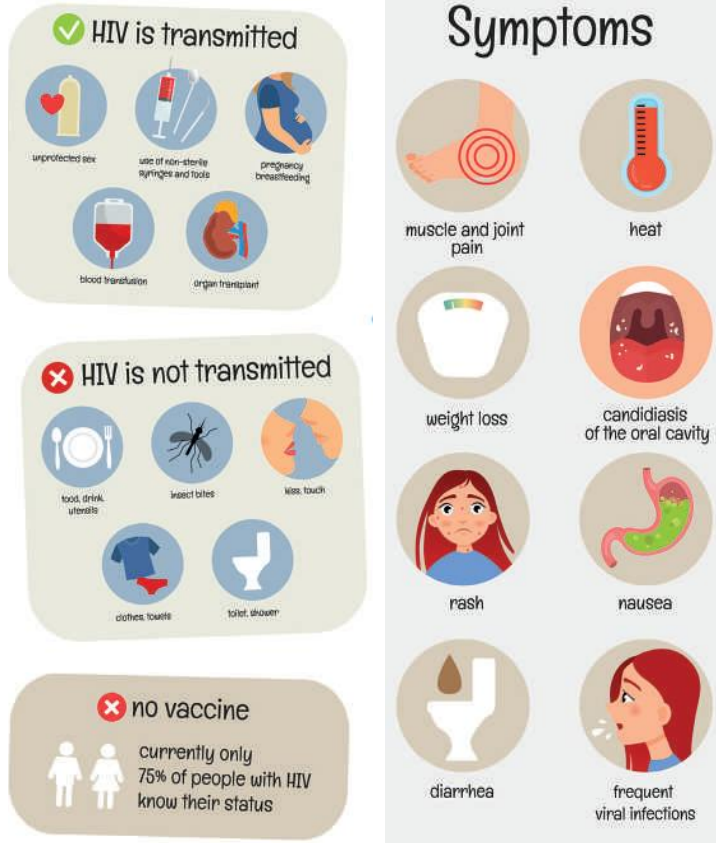


Figure 28.6: Transmission and symptoms of HIV AIDS

There is no cure for HIV infection. However, with access to effective HIV prevention, diagnosis, treatment and care, HIV infection has become a manageable chronic health condition, enabling people living with HIV to lead long and healthy lives.

HPV (HUMAN PAPILLOMAVIRUS)

HPV usually shows no symptoms and goes away by itself, but can sometimes cause serious illness.

HPV is responsible for:

- almost all cases of genital warts and cervical cancer
- 90% of anal cancers
- 78% of vaginal cancers
- 25% of vulvar cancers
- 50% of penile cancers
- 60% of oropharyngeal cancers (cancers of the back of the throat, including the base of the tongue and tonsils).

The human papillomavirus (HPV) vaccine used in Australia is called Gardasil ®9 and protects against nine types of HPV that are responsible for most HPV-related illnesses.

The HPV vaccine is offered to all Australian children (females and males) in Year 7 for free through the Secondary School Immunisation Program as part of the National Immunisation Program.

Figure 28.7: HPV vaccine available to all children aged 12–13 in Australia

HEPATITIS

There are 5 types of hepatitis caused by viruses, labelled A–E.

Vaccines can protect you against hepatitis A, B and D.

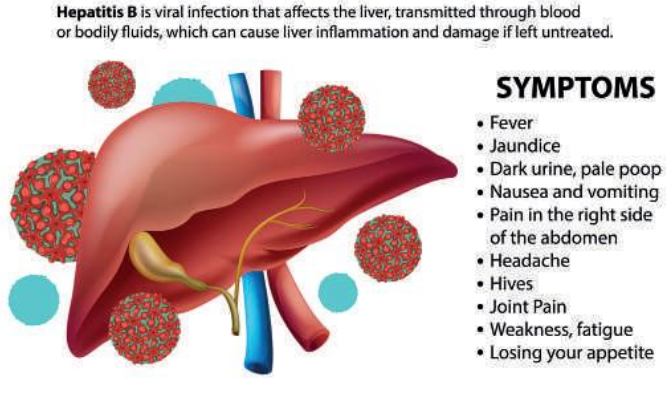


Figure 28.8: Symptoms of Hepatitis B

More information on sexually transmitted diseases can be found at:

https://sti.guidelines.org.au/sexually-transmissible-infections/

Question 1

Which of these STIs can be cured with antibiotics? (1 mark)

(a) Herpes
(b) Chlamydia
(c) Genital warts
(d) HPV

Question 2

Herpes can be transmitted by all of these methods, except: (1 mark)

(a) kissing.
(b) from sharing the same lip-gloss.
(c) sexual intercourse.
(d) from a toilet seat.

Question 3

A vaccine is available to Australian children at the age of 12–13 for which STI? (1 mark)

(a) HPV
(b) HIV
(c) Herpes C and D
(d) FDS

Question 4

As long as a person has no symptoms of STIs, (1 mark)

(a) they cannot pass it on.
(b) they don't need treatment.
(c) they won't get it again.
(d) none of the above.

Question 5

Which of the following groups of STIs can be treated with antibiotics? Those caused by: (1 mark)

(a) bacteria and viruses.
(b) viruses and fungi.
(c) bacteria and some parasites.
(d) viruses and some fungi.

Question 6

The main organ of the body effected by hepatitis is: (1 mark)

(a) the lungs.
(b) the heart.
(c) the liver.
(d) the skin.

Question 7

Left untreated, which STI can lead to deafness and death in later stages? (1 mark)

(a) Chlamydia
(b) HIV
(c) HPV
(d) Syphilis

Question 8

Which of the following is not a symptom an STI? (1 mark)

(a) Coughing and sneezing
(b) Lumps, sores or warts near the mouth, anus or vagina
(c) Smelly secretions from penis or vagina
(d) Painful urination

Question 9

Select ALL of the true statements below.

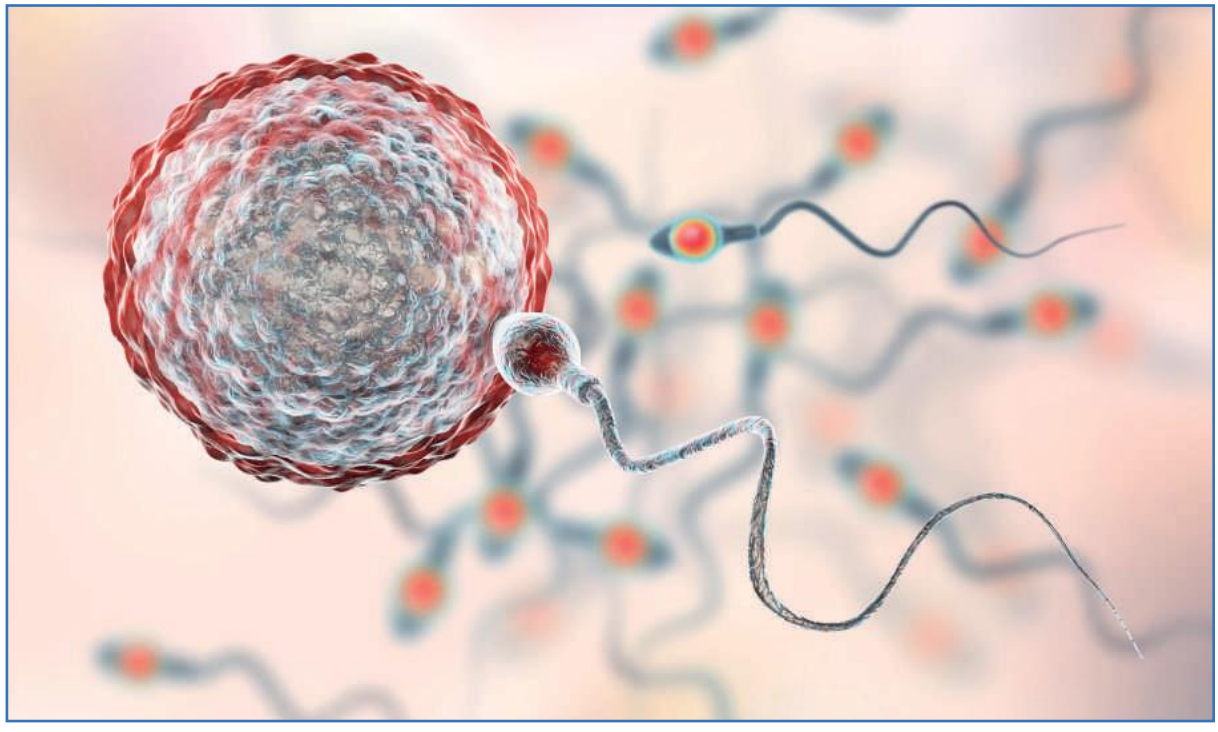
(a) All STIs can be cured.
(b) All STIs have symptoms.
(c) Some STIs result in infertility. (1 mark)
(d) Some STIs can be cured. (1 mark)
(e) Some STIs can result in infertility. (1 mark)
(f) Some STIs can result in death. (1 mark)

Question 10

Indicate whether these statements are true or false. (1 mark for each)

(a) It is normal for women to have some vaginal discharge. T / F
(b) Once you have had an STI, you can't get it again. T / F
(c) HIV can be transmitted through unscreened blood transfusions. T / F
(d) Most STIs go away without treatment, if people wait long enough. T / F
(e) The contraceptive pill provides protection against some STIs. T / F
(f) A pregnant woman with a STI can pass it onto her baby. T / F
(g) A person can have an STI and not know it. T / F
(h) All itching in the genital area is caused by an STI. T / F

29 CONCEPTION AND IMPLANTATION



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• For the establishment of a pregnancy, conception requires the union of viable sperm and ovum at the optimal time in the ovarian cycle.				
• Greater understanding of the menstrual cycle, conception and implantation has produced improved methods of the establishment of a pregnancy, along with advancements in contraceptive methods; both have ethical considerations.				

Conception is the fertilisation of the ovum by a sperm.

Pregnancy is not established until the fertilised ovum is implanted into the endometrium.

CONCEPTION

Conception occurs in the upper third of the fallopian tube.

A structure called the acromere on the head of the sperm contains enzymes to break through the corona radiata and penetrate the zona pellucida.

Task 29.1: Label the zona pellucida and the corona radiata in Figure 29.1.

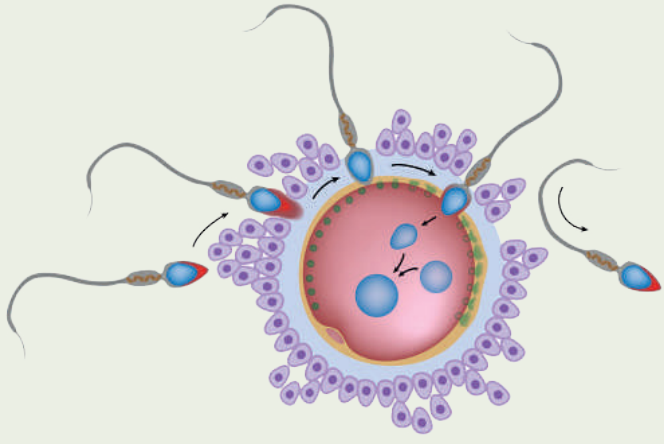


Figure 29.1: Fertilisation of the ovum

Conception can occur when a viable ovum and sperm are present together in the fallopian tube. The ovum is usually viable for 12–24 hours after ovulation. Sperm can survive up to 5 days in the female reproductive tract.

Conception can therefore take place if sexual intercourse has occurred any time between day 9 and about day 16–17 of the menstrual cycle. This is dependent on when ovulation occurs – five days before and at least 2 days after intercourse.

SEX DETERMINATION

The sex of the offspring is determined at fertilisation.

The female genotype is XX. The male genotype is XY.

Sex is controlled by the inheritance of the X and Y chromosomes carried in the gametes.

Male gametes can carry either X or Y chromosomes.

Female gametes only carry X chromosomes.

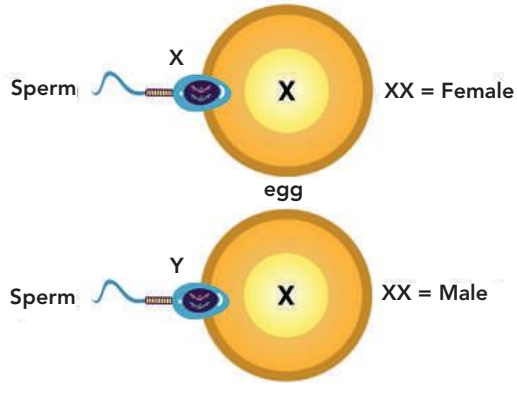


Figure 29.2: Genotypes of males and females

IMPLANTATION

The zona pellucida prevents polyspermy by reacting immediately to the breach in the membrane by a sperm, so other sperm cannot enter.

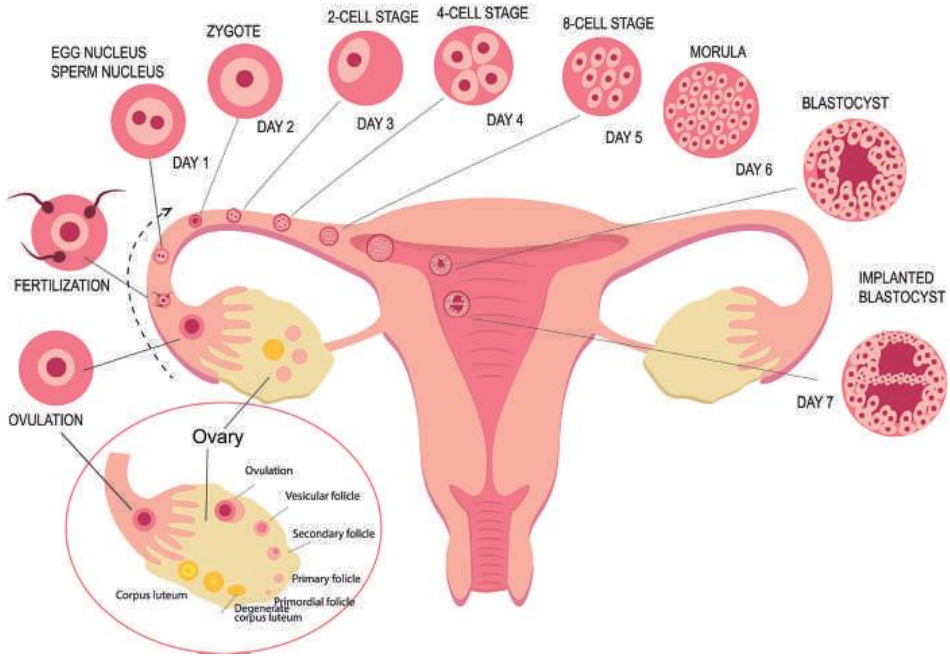


Figure 29.3: Ovulation and fertilisation process

By the time the fertilised ovum reaches the uterus, it has undergone many mitotic cell divisions to form the different stages:

- Morula – a solid ball of cells
- Blastocyst – formed when a fluid filled cavity appears in the morula between the cells of the inner cell mass and the enveloping layer; cell differentiation has started to occur.

The inner cell mass will become the foetus and the enveloping layers will form the placenta and amnion.

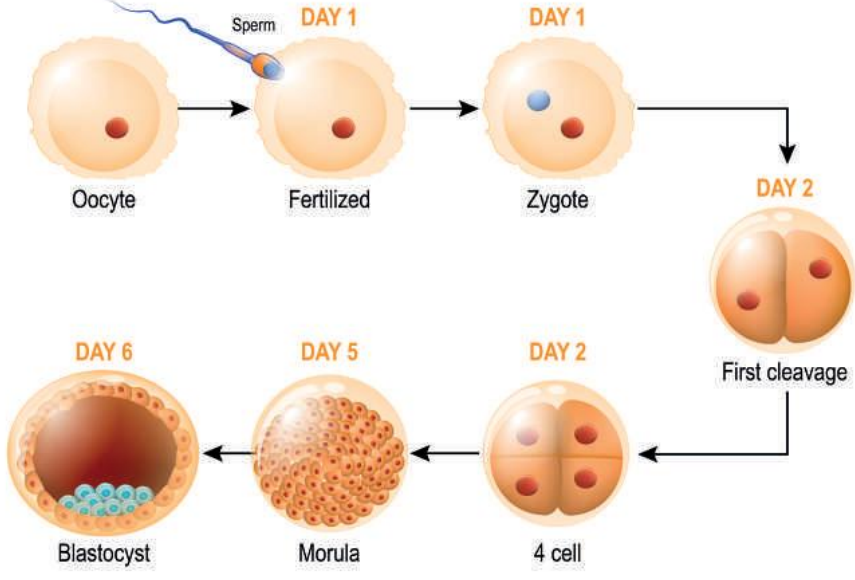


Figure 29.4: Development of the embryo

It is at the blastocyst stage that implantation occurs. Note, the zona pellucida is still present.

A blastocyst, at around five days after fertilisation, 'hatches' out of its protective the zona pellucida. The removal of the zona pellucida is a critical step that is necessary in order for the blastocyst to communicate and have cell-to-cell interaction with the uterine lining and eventually implant securely.

In humans, implantation begins at the end of week 1, with most successful human pregnancies the blastocyst implants 8 to 10 days after ovulation. Early pregnancy loss increases with later implantation.

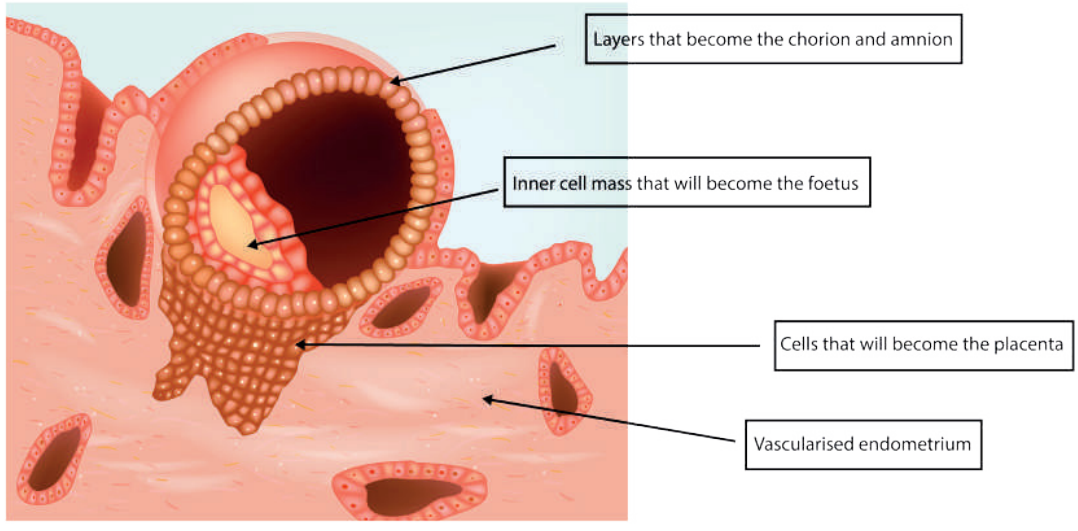


Figure 29.5: Implanting blastocyst

Implantation causes the developing placenta to produce human chorionic gonadotrophic hormone (hCG).

Pregnancy test kits test are available to test for the presence of this hormone in blood 10–11 days after conception and slightly later when testing urine. This is before the next menstrual flow is expected.

hCG helps thicken the uterine lining to support a growing embryo and tells the body to stop menstruation. hCG levels rise after conception and continue to rise until about 10 weeks in pregnancy.

Question 1

Explain how conception can occur without resulting in pregnancy. (2 marks)

Question 2

Describe the structures of the sperm and ovum that promote conception. (2 marks)

Question 3

Explain why so many sperm are required for conception to occur. (4 marks)

Question 4

Explain the problem of conception occurring when the ovum reaches the uterus. (2 marks)

Question 5

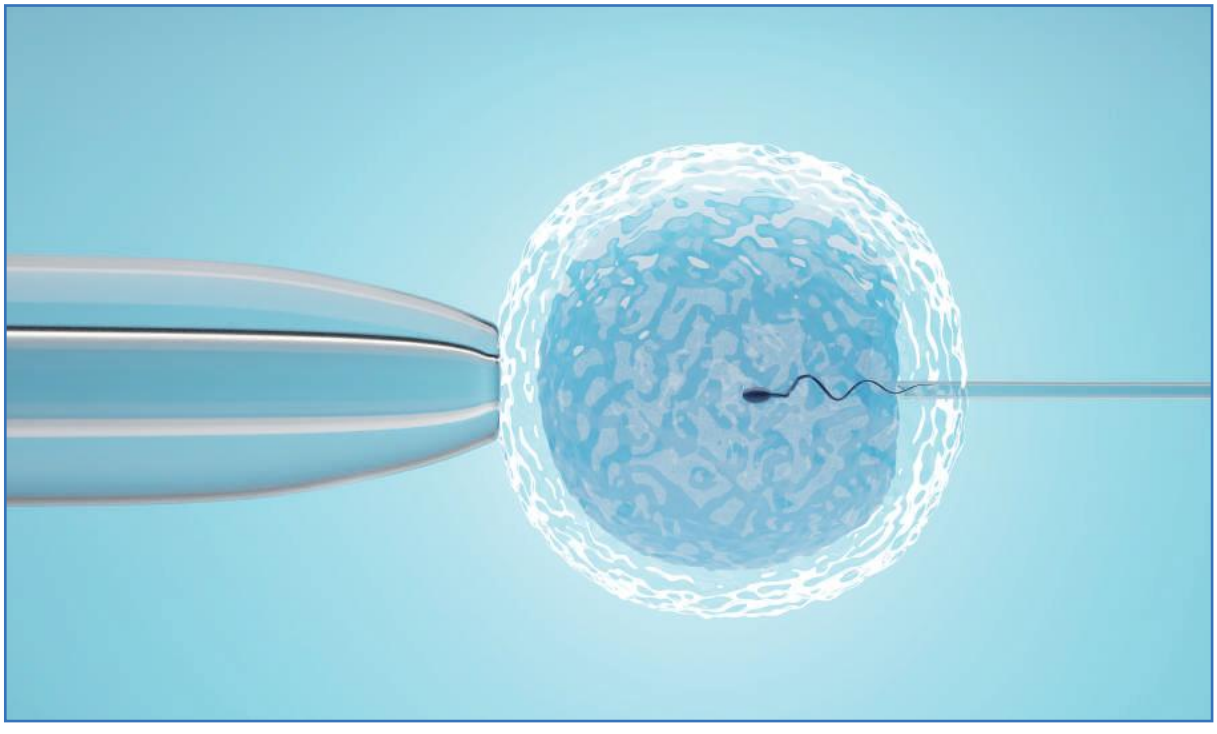
The zona pellucida can breakdown at about the 2 or 4 cell stage. Describe a possible outcome of this. (2 marks)

Question 6

Look at Figure 29.3 carefully. The morula does not have differentiated cells but the blastocyst does. Look carefully at the structure of each and suggest why the differentiation in the blastocyst produces different layers at different locations. (2 marks)

ASSISTED REPRODUCTIVE TECHNOLOGIES (ART)

30



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• There are a variety of assisted reproductive technologies to help overcome infertility problems, but each has its limitations, risks and benefits.				

These technologies refer to fertility treatments and procedures that can help overcome difficulties a person or couple is experiencing in conceiving and establishing a pregnancy.

ART techniques involve the manipulation of eggs, sperm, or embryos to increase the likelihood of a successful pregnancy

Infertility is when people cannot conceive after a period of regular sexual intercourse without the use of birth control. Research indicates that worldwide, 8–12% of couples experience fertility problems, and 40–50% of cases may stem from factors that affect males.

OVARIAN INDUCTION

- Used when ovulation is not occurring or is not regular.
- Hormones are used to stimulate the development and maturation of the ovarian follicles and to trigger ovulation.

Risks:

- More than one ovum may be released, increasing the chance of multiple implantations
- Ovarian hyperstimulation can cause nausea, vomiting, diarrhea, shortness of breath, weight gain and dehydration.

Benefits:

- Regulating menstrual cycle to time intercourse with ovulation for highest chance of conception.

ARTIFICIAL INSEMINATION OR INTRAUTERINE INSEMINATION

- Sperm is collected and assessed for viability.
- The menstrual cycle is controlled and monitored.
- The time of insemination is chosen when ovulation is imminent.
- Sperm is delivered directly to the cervix or into the uterus for the sperm to move into the fallopian tube where an ovum is, hopefully, present.
- Fertilisation takes place and the rest follows the natural course of events.

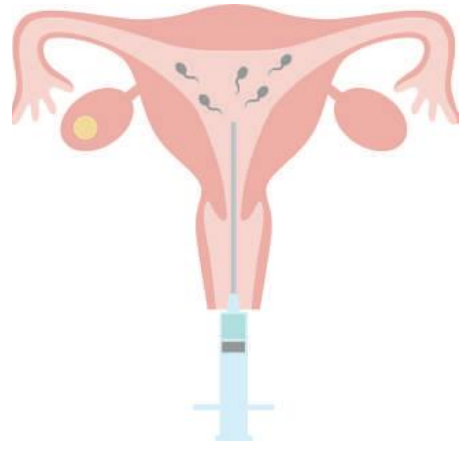


Figure 30.1: Artificial insemination

Risks:

- Almost negligible if hormone treatment is not involved.

Benefits:

- Overcomes male infertility such as low sperm count or abnormal sperm
- Can be used with sperm donations.

IN VITRO FERTILISATION (IVF)

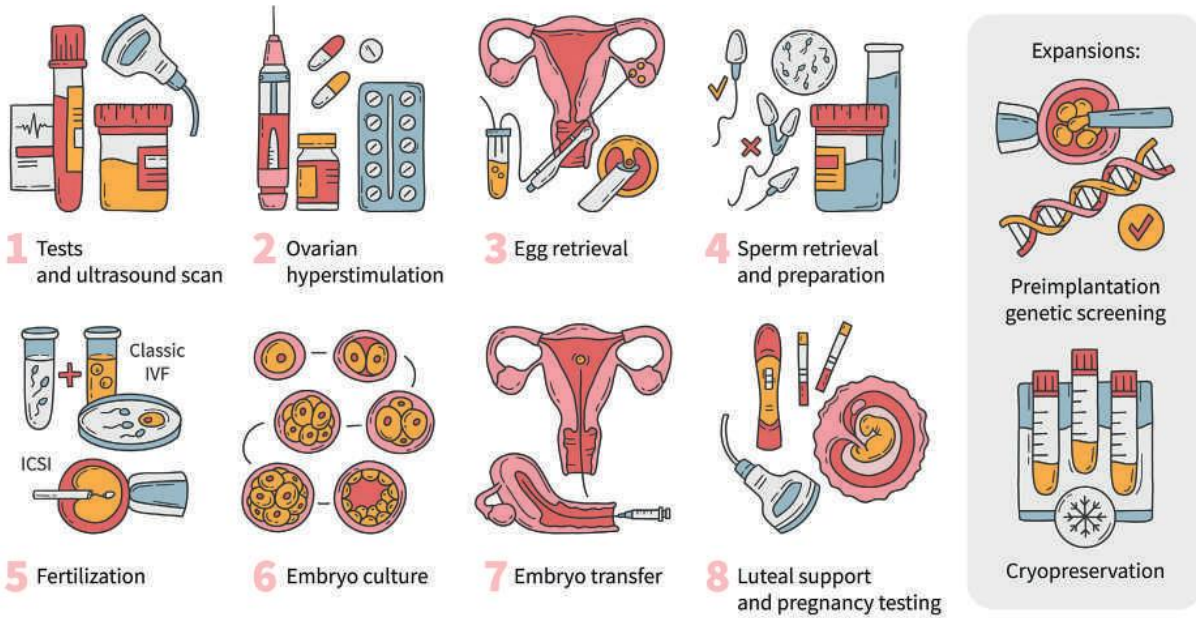


Figure 30.2: Steps in IVF procedure

In vitro fertilisation occurs outside the body, usually in a laboratory glassware and the resulting embryo is implanted into the prepared uterus.

In vitro – from the Latin, in glass.

Benefits:

- bypasses blocked fallopian tubes
- overcomes, to some extent, male infertility or abnormal sperm
- gametes can be screened for genetic conditions such as Down's syndrome and cystic fibrosis.

Success rates:

- 52% for females aged 35 or younger
- 7.6% for females aged 40 and over.

Risks:

- multiple pregnancy – two or more embryos implanting at a time
- side effects from fertility drugs, such as ovarian hyperstimulation syndrome.

INTRACYTOPLASMIC SPERM INJECTION (ICSI)

ICSI follows the same process as IVF, except in ICSI the embryologist uses a microscope to select a single sperm that is injected into the egg to hopefully achieve fertilisation.

ICSI is sperm selection procedure used to overcome male factor infertility, including:

- low sperm count,
- low sperm motility (movement)
- poor sperm morphology (shape)
- problems with ejaculation.

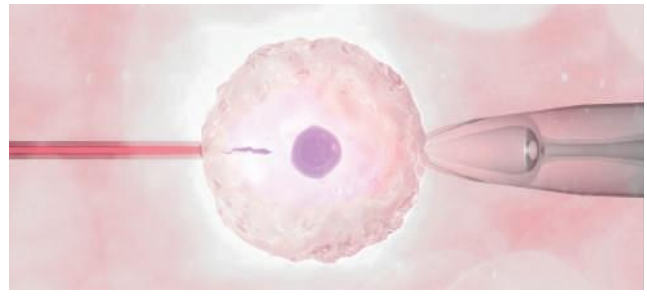


Figure 30.3: ICSI process

Risks:

- small increase in the risk of still birth, premature birth, low birth weight and multiple birth
- about 20% risk of miscarriage.

Benefits:

- overcomes male fertility problems
- genetic screening available
- use of donor eggs and sperm possible.

Female fertility problems

- Increased age and the onset of menopause reduces the production of viable ova and the chances of establishing a pregnancy.
- Irregular menstrual cycles interfere with the development of mature ova and ovulation.
- Blocked fallopian tubes prevent the passage of the ovum and sperm so there is a much reduced chance of conception taking place.
- Polycystic ovaries is a problem with hormones. Many small sacs of fluid develop along the outer edge of the ovary called cysts. The small fluid-filled cysts contain immature eggs. The follicles fail to regularly release eggs.

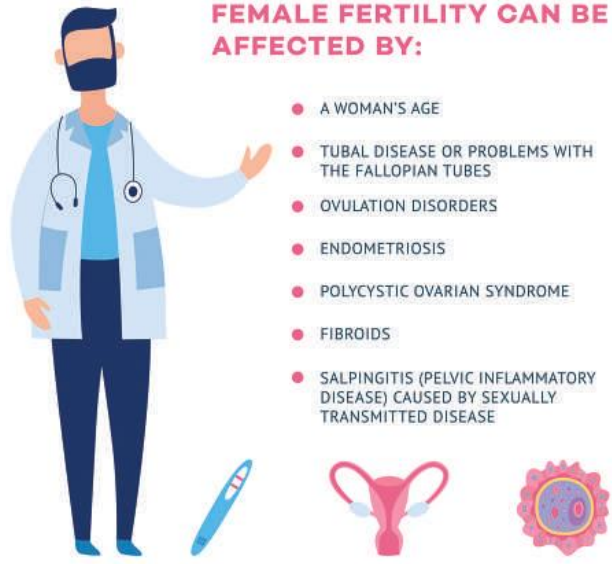


Figure 30.4: Female infertility problems

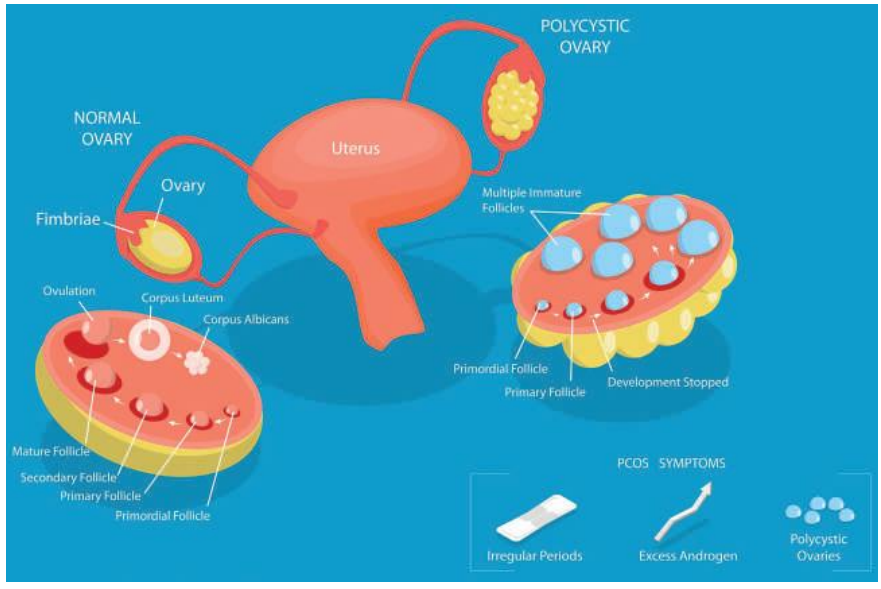


Figure 30.5: Polycystic ovaries

- **Fibroids** are non-cancerous tumours in the uterus can block the:
  - passage of sperm to the fallopian tube
  - fertilised ovum to the uterus
  - implantation of the embryo.

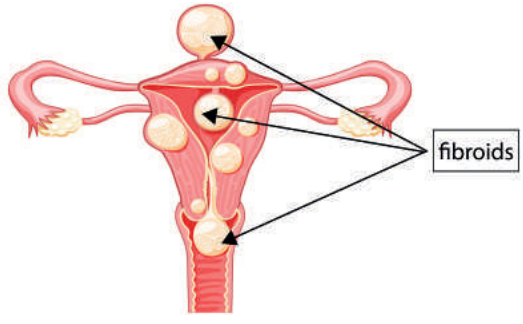


Figure 30.6: Common locations of fibroids in and around the uterus

- **Endometriosis** is when body tissue like the lining of the uterus, called the endometrium, grows in other parts of the body e.g. the outside of the uterus, the ovaries, large intestine and the abdominal cavity. The tissue responds to the hormone oestrogen released from the ovaries. During menstruation, the tissue outside the uterus also breaks down which can lead to pain, inflammation and scarring, causing organs to stick together (known as adhesions). Internal abdominal movements can then cause discomfort or pain.

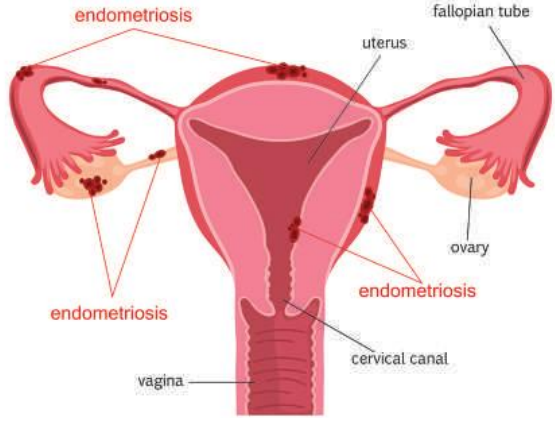


Figure 30.7: Endometriosis

Male fertility problems

Male infertility can result from anatomical or genetic abnormalities, systemic or neurological diseases, infections, trauma, injury caused by medical or surgical treatment, gonadotoxins (chemicals toxic to the gonads) and development of sperm antibodies.

A lower than normal sperm count (fewer than 15 million sperm per millilitre of semen or a total sperm count of less than 39 million per ejaculate).

Figure 30.8: Male fertility problems

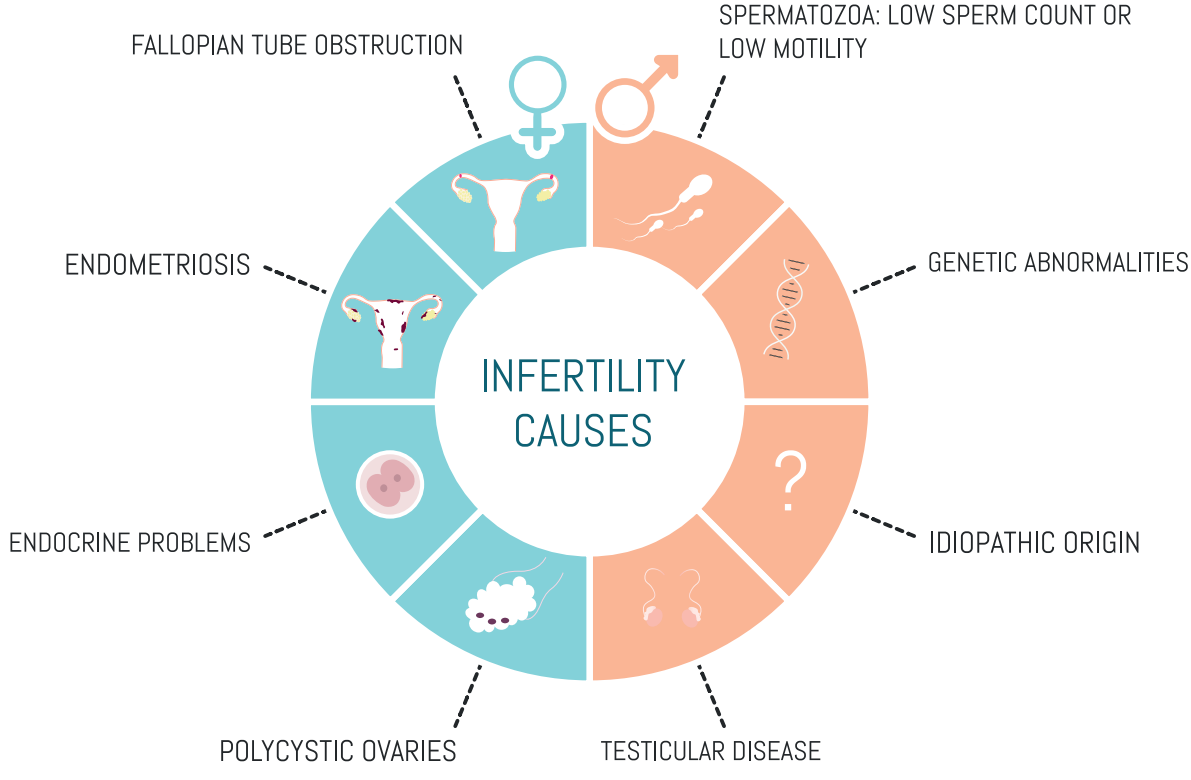


Figure 30.9: Overview of fertility problems in males and females

Questions

(20 marks)

	Statement	True	False
1	Men do not experience an age related decrease in fertility.		
2	Men and women are equally likely to have fertility problems.		
3	In vitro fertilisation (IVF) is a simple, cost-effective procedure for infertile couples.		
4	Fertility in women is impaired by obesity.		
5	For women over 30, overall health and fitness levels are better indicators of fertility than age.		
6	Prior to a women reaching menopause, the assisted reproductive technologies (e.g., in vitro fertilisation, IVF) can help most women to have a baby using her own eggs.		
7	A woman's fertility starts to decline in her late 20s.		
8	In most cases, couples with fertility problems need to undergo complicated, high-tech procedures such as in vitro fertilisation.		
9	Couples younger than 35 years of age are considered infertile if they have not been able to conceive naturally for 12 months after discontinuing the use of contraceptives.		
10	Smoking doesn't affect fertility in women.		

31 GENETIC TESTING AND EMBRYONIC SCREENING

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• There are a range of techniques available to screen embryos before implantation or during early development, including blood tests, ultrasound, amniocentesis and chorionic villi sampling.				
• The use of genetic screening to assess the risk of inherited disorders has implicit ethical considerations.				

The following table is a summary of some of the antenatal tests routinely offered during pregnancy in Australia. Not all pregnant women need to do all the tests.

Name	Type of test	What does it check for?	When is it done?
Infectious disease screen	Blood test	Immunity to infections such as hepatitis, HIV and rubella	First antenatal visit
Blood group + antibodies	Blood test	Blood group	First antenatal visit
Full blood count	Blood test	Anaemia	First antenatal visit
Vitamin D level	Blood test	Vitamin D deficiency	First antenatal visit
Urine culture	Urine test	Infection	First antenatal visit May be repeated during pregnancy
Dating scan	Ultrasound scan	Estimated due date	8–14 weeks
Nuchal translucency test	Ultrasound scan	Screens for genetic abnormalities	11–13 weeks
Combined first-trimester screen (CFTS)	Ultrasound scan + blood test	Screens for genetic abnormalities	11–14 weeks
Non-invasive prenatal testing (NIPT)	Blood test	Screens for genetic abnormalities	From 10 weeks
Chorionic villus sampling (CVS)	Procedure	Diagnoses genetic abnormalities	From 11 weeks
Amniocentesis	Procedure	Diagnoses genetic abnormalities	From 15 weeks
Morphology scan	Ultrasound scan	Foetal growth and development Position of the placenta	18–20 weeks
Gestational diabetes screening	Blood test	Gestational diabetes	24–28 weeks
Group B strep screen	Vaginal-rectal swab	Group B strep	30–36 weeks

GENETIC TESTING

Genetic testing involves analysis of a person's DNA to see if they carry alleles that cause genetic disorders. It can be done at any stage in a person's life.

- **Pre-implantation genetic diagnosis** (PGD) is used on embryos before implantation. Once the embryos have reached the eight-cell stage, one cell is removed. The cell is tested for the disorder-causing alleles and chromosomal abnormalities. Embryos that don't contain the disorder allele are implanted into the uterus.
    - PGT-A: Preimplantation genetic testing for aneuploidy. This test is to make sure that the embryo has the correct number of chromosomes.
    - PGT-SR: A test to verify if there are any structural chromosome rearrangements. When a parent has a modified chromosome structure, the test can detect missing or excessive genetic material in an embryo.

- PGT-M: PGT for single-gene disorders. Some known inherited disorders are caused by a single gene, and this type of embryo test focuses on those disorders (DNA sequencing used).

- **Antenatal testing** is used to analyse an individual's DNA or chromosomes before they are born. This testing is offered to couples who may have an increased risk of producing a baby with an inherited disorder, but it can't detect all the risks of inherited disorders.
- **Neonatal testing** known as the new born blood spot test involves analysing a sample of blood that is taken from pricking a baby's heel. It detects genetic disorders in order to treat them early.

Genetic testing (PGT) has markedly improved clinical pregnancy outcomes for carriers of gene mutations or chromosomal structural rearrangements by the selection of embryos free of disease-causing genes and chromosome abnormalities.

Genetic testing enables chromosomally healthy embryos or those unaffected by a specific disorder to be selected for transfer during an IVF cycle, increasing the chance of a healthy baby.

KARYOTYPING

A karyotype is an individual's complete set of chromosomes. The term also refers to a laboratory-produced image of a person's chromosomes isolated from an individual cell at prophase of mitosis and arranged in numerical order. A karyotype may be used to look for abnormalities in chromosome number or structure.

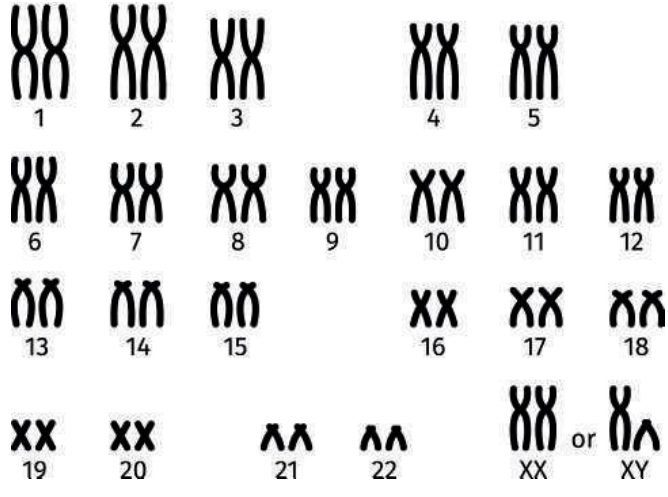


Figure 31.1: Normal human karyotype

The karyotypes below in **Figures 31.2–31.7** show some of the chromosomal aberrations that can be detected through genetic screening.

Task 31.1: For each of the karyotypes shown in Figures 31.2–31.7, circle and describe the chromosomal aberration and the sex of the individual in the space next to each karotype.

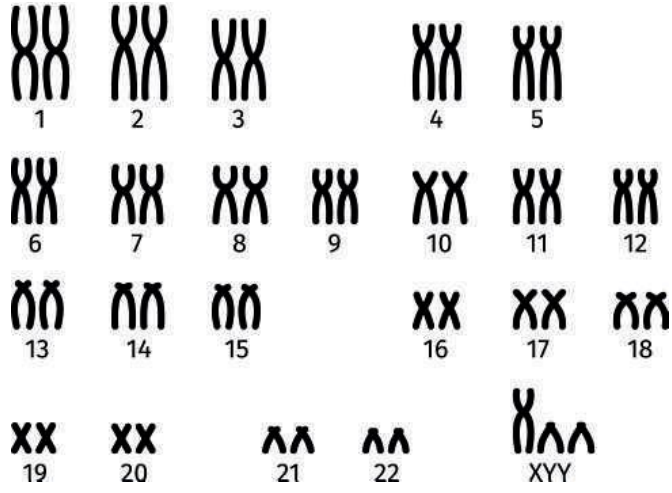


Figure 31.2: Jacob's syndrome

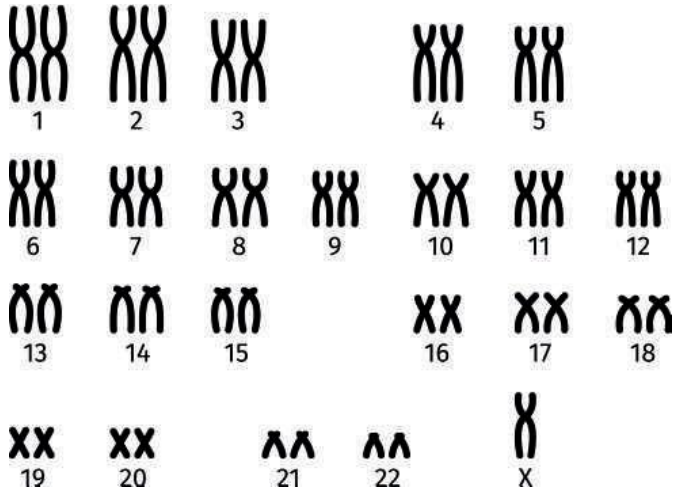


Figure 31.3: Turner's syndrome

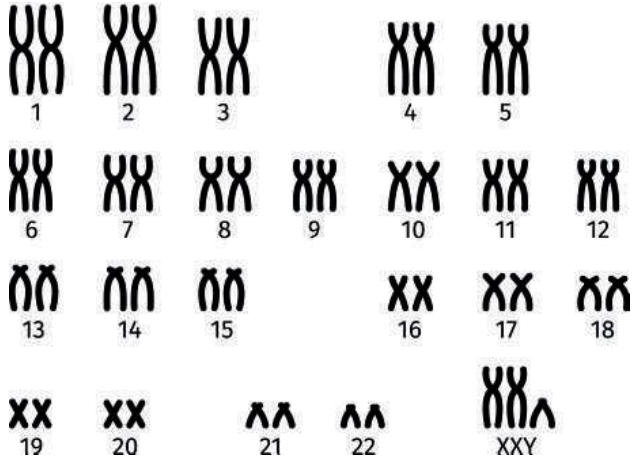


Figure 31.4: Klinefelter's syndrome

The following conditions can be detected using an ultrasound scan with a blood test that is carried out between 10 and 14 weeks of pregnancy.

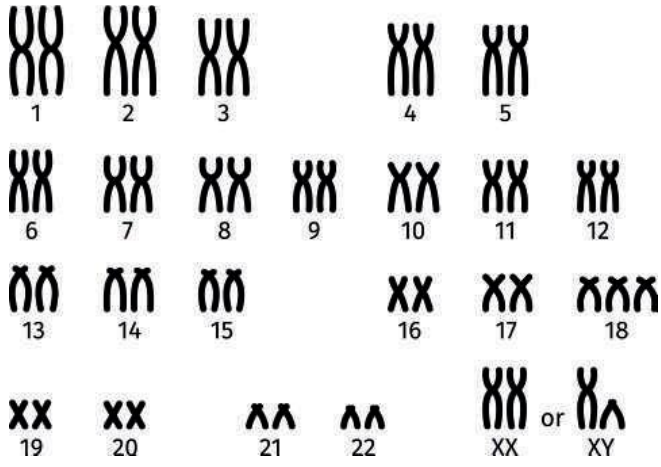


Figure 31.5: Edward's syndrome

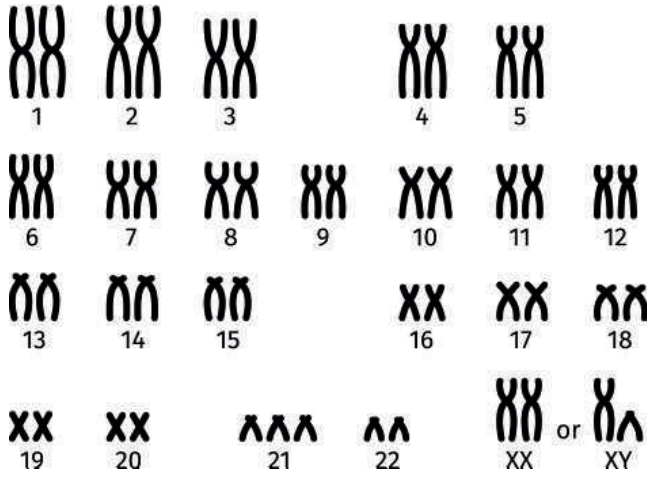


Figure 31.6: Down's syndrome

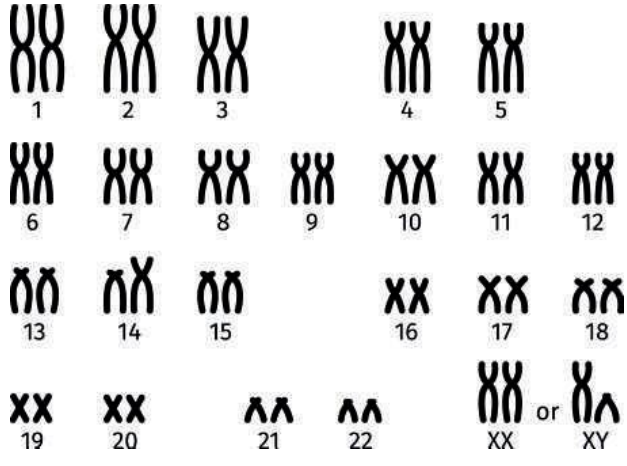


Figure 31.7: Robertson's translocation

These aberrations are caused by errors in the separation processes in meiosis.

EMBRYONIC SCREENING

ULTRASOUND

High frequency sound waves are used to produce an image of the foetus in utero.

This is used to make body measurements to monitor developmental progress and determine the location of the placenta.

It can also be used to determine the sex of the foetus.

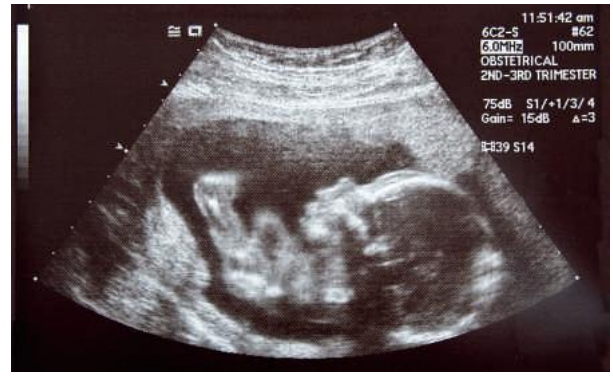


Figure 31.8: Ultrasound image of a foetus

BLOOD TESTS

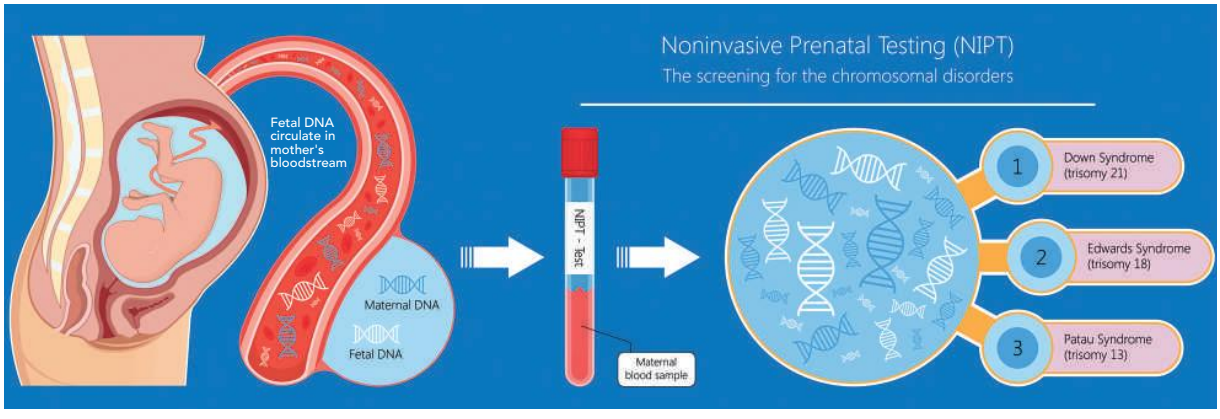


Figure 31.9: Non-invasive prenatal testing NIPT screening genetic disorders using blood

AMNIOCENTESIS

Ultrasound is used to guide the needle to collect a sample of amniotic fluid that surrounds the foetus. This fluid contain cells from the foetus.

The cells can be cultured and screened for genetic aberrations, provide information on neural tube defects e.g. spina bifida, look for infections and test for foetal lung development later in pregnancy.

The fluid undergoes chemical analysis to check if the contents all are within the accepted ranges.

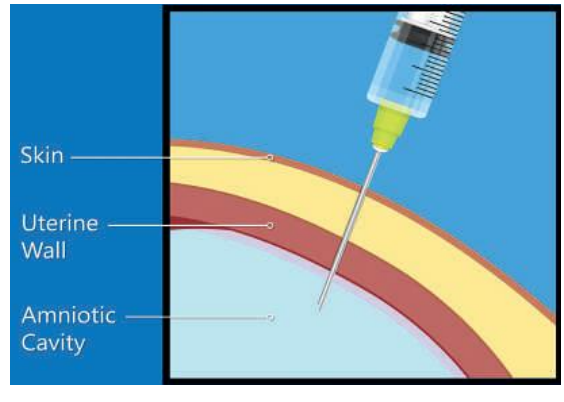


Figure 31.10: Amniocentesis

CHORIONIC VILLI SAMPLING

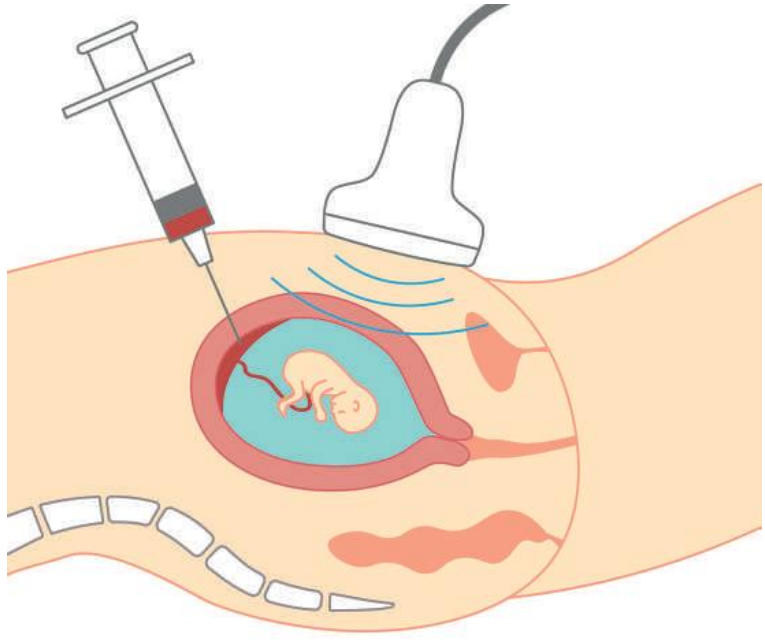


Figure 31.11: Chorionic villi sampling

Task 31.2: Label the following in Figure 31.11: foetus, umbilical cord, syringe, uterine wall, amniotic fluid, placenta, cervix, vagina bladder, ultrasound machine, spine, large intestine

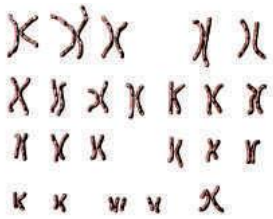
Ultrasound is used to guide the needle to the placenta where a sample is taken. It will contain cells from the mother and the foetus. The sample is tested for chromosomal abnormalities and certain other genetic problems.

Reasons for having a CVS are:

- previously affected child or a family history of a genetic disease, chromosomal abnormalities, or metabolic disorder
- maternal age over 35 years by the pregnancy due date
- risk of a sex-linked genetic disease
- previous ultrasound with questionable or abnormal findings.

Question 1

Describe the phenotype of the person with this karyotype. (3 marks)



Question 2

Describe the differences between amniocentesis and chorionic villi sampling. (2 marks)

Question 3

Outline the risks of chorionic villi sampling. (4 marks)

Question 4

List three reasons why screening tests are done. (5 marks)

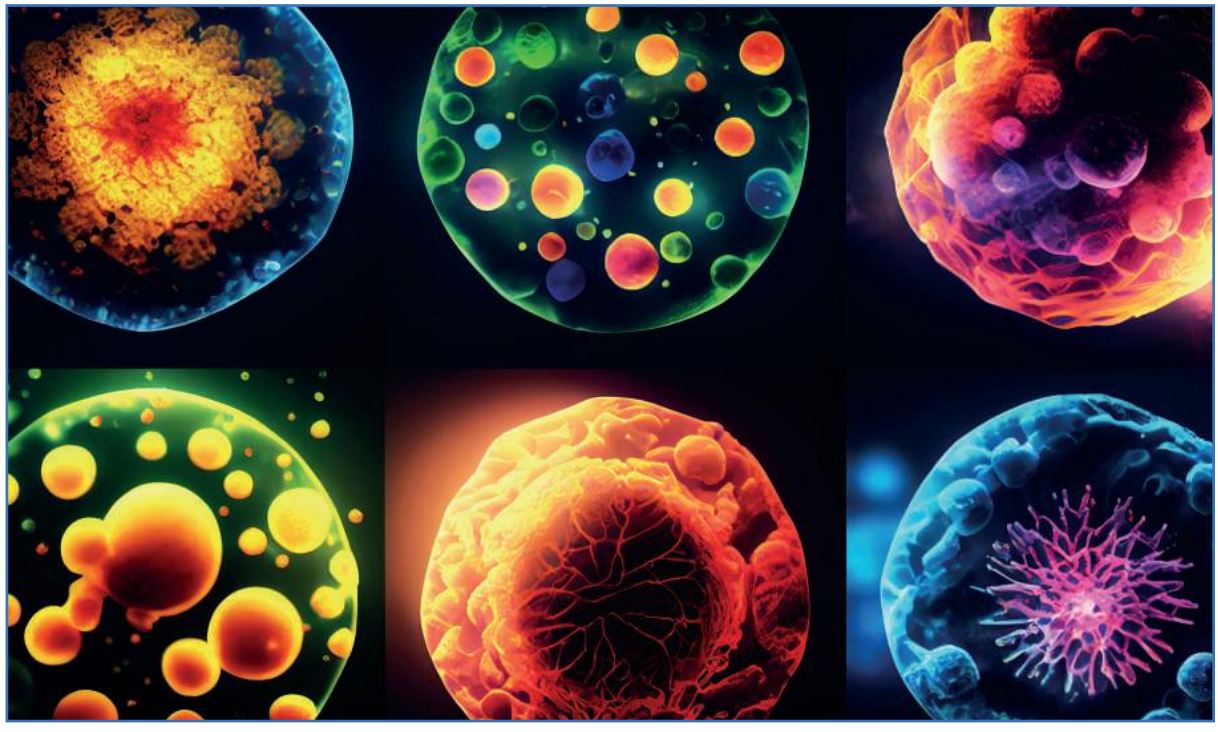
Question 5

What are the possible actions parents could take if a screening test is positive for a genetic aberration? (2 marks)

Question 6

Explain how the genotype of Down's syndrome occurs. (2 marks)

32 CELL POTENCY AND CANCER



Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Stem cells have the ability to divide by mitosis and differentiate into many different tissues, depending on the level of cell potency.				
• Uncontrolled division of cells can result in the development of tumors/cancers.				
• New technologies, including the cervical screening test, breast screening and blood tests for prostate cancer, have made early detection of many cancers possible.				

The human body starts as one cell, the zygote (fertilised ovum).

From this one cell is derived all the cells and tissues of the body.

It is the ultimate stem cell, cells that can differentiate into various cell types.

Not all stem cells can differentiate into all other types of cells.

There are different levels of **potency** determined by the extent of the differentiation of daughter cells.

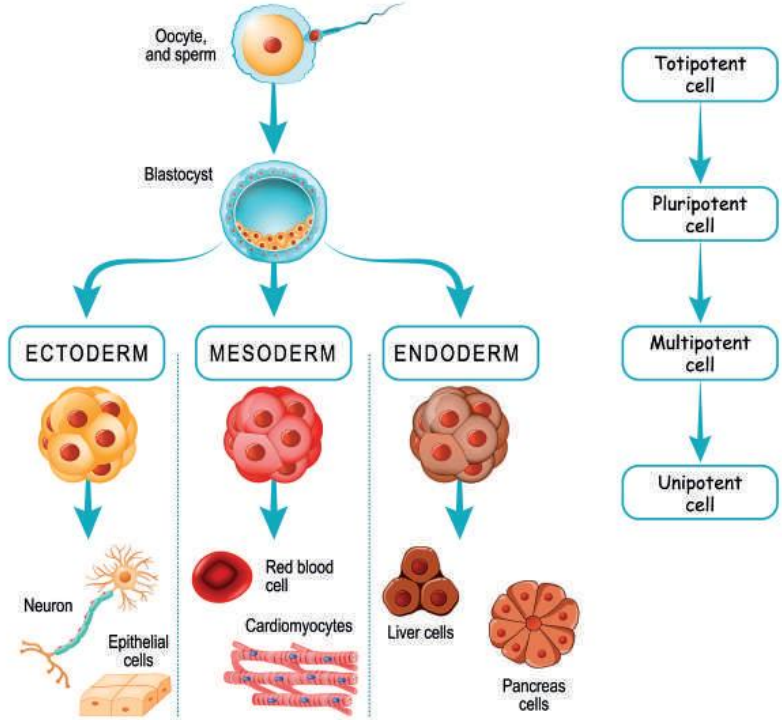


Figure 32.1: Levels of cell potency

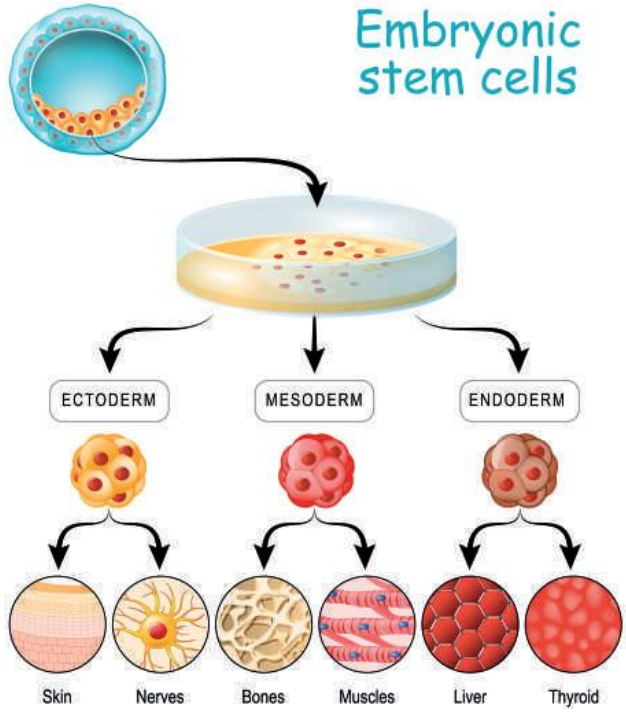


Figure 32.2: Pluripotency of cells from the blastocyst

The zygote produces the cells of the inner cell mass, which will become the embryo and the placental membranes. The cells in the inner cell mass produce all the cell types required in the developing embryo (not the placental membranes). This is the difference between totipotent and pluripotent cells.

The cells of the blastocyst gradually lose this level of potency as it develops into different layers of cells that will become the ectoderm, mesoderm and endoderm, limiting the different types of cells into which they can differentiate.

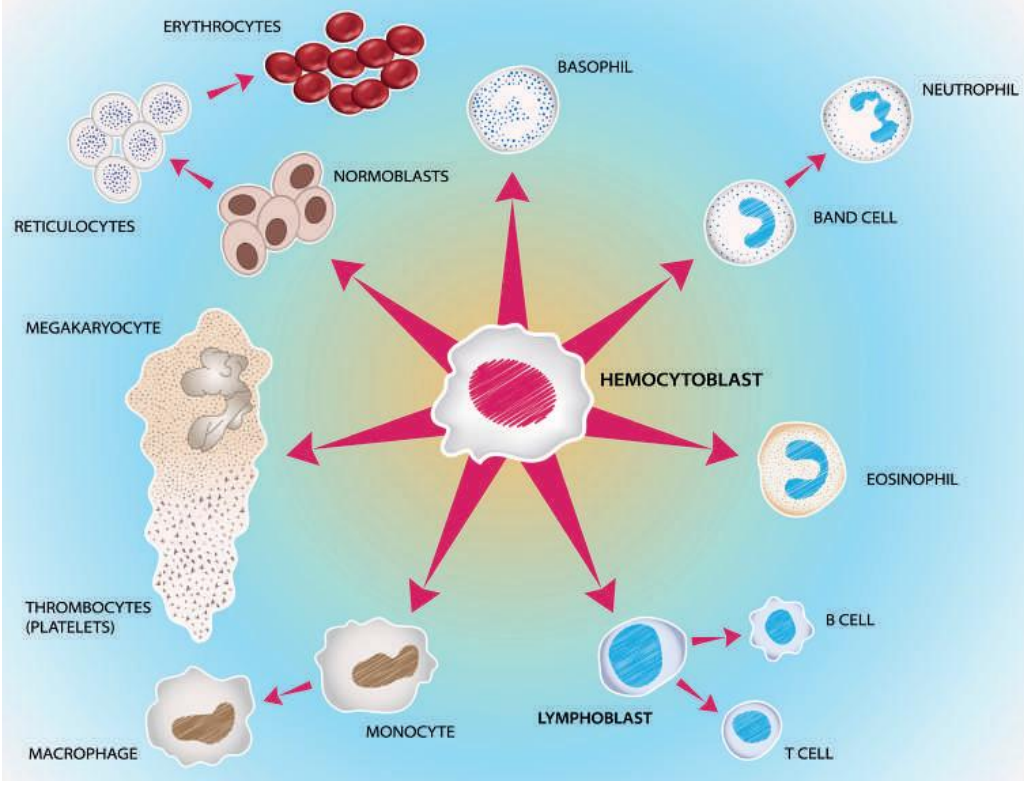


Figure 32.3: Multipotency of hemocytoblast

Hemocytoblasts are generalised stem cells found in the bone marrow, from which all blood cells form, including both erythrocytes and leukocytes. Differentiation is limited to types of blood cells.

USES OF STEM CELLS

Stem cells are cells that can develop into different types of cells in the body. They have various uses.

Natural and scientific uses of stem cells:

- Repairing and renewing damaged or diseased tissues and organs
- Replacing cells that are lost due to aging, injury, or disease
- Scientific research
- Serving as a way to fight some types of cancer and blood-related diseases
- Researching causes and treatments of diseases and genetic defects
- Testing new drugs for safety and effectiveness

Therapeutic use of stem cells

- Stem cells can be used to replace damaged / diseased cells with healthy ones.
- Stem cells can be harvested from embryos, umbilical cord blood or certain adult tissues (e.g. bone marrow).
- Biochemical solutions are used to trigger the differentiation of stem cells.
- Cells are surgically implanted into patient's own tissue and the host immune system is suppressed to prevent rejection of cells.
- New cells must be monitored to ensure they do not become cancerous.

Sources of stem cells:

- **Embryonic stem cells** come from embryos that are 3 to 5 days old. At this stage, an embryo is called a blastocyst and has about 150 cells. These cells of the inner cell mass are pluripotent. This versatility allows embryonic stem cells to be used to regenerate or repair diseased tissue and organs.
- **Adult stem cells** are found in small numbers in most adult tissues, such as bone marrow or fat. Compared with embryonic stem cells, adult stem cells have a more limited ability to give rise to various cells of the body. Adult stem cells only grow in specific parts of the body including: bone marrow, breasts, intestines, fat tissue, brain, nose, hair follicles and testes. These cells can only differentiate into the cells that grow in that region.
- **Adult cells altered to have properties of embryonic stem cells** are transformed regular adult cells into stem cells using genetic reprogramming. By altering the genes in the adult cells, the cells can be reprogrammed to act similarly to embryonic stem cells.
- **Perinatal stem cells** are stem cells in amniotic fluid and umbilical cord blood. These stem cells have the ability to change into specialised cells. Umbilical cord blood can be stored for later use to treat the child, if required.

Adult stem cells in humans have been used to treat diseases like leukaemia and other cancers of the blood and bone marrow by transplants. Theoretically, the use of stem cells could treat a large range of diseases like type 1 diabetes, Parkinson's disease, and brain and spinal cord injuries. However this is proving difficult to do. Because adult stem cells can only differentiate into one or several cell types they are not as useful as embryonic stem cells. However the use of embryonic stem cells has clinical, ethical and social issues associated with them.

CANCER

When cells in an area of the body divide without control, excess tissue forms that is called a tumour. Tumors that do not spread to other parts of the body are called **benign** cancers or tumors.

Uncontrolled cell growths that spread to other parts of the body are called **malignant** tumors.

Metastases most commonly develop when cancer cells break away from the main tumour and enter the body's bloodstream or lymphatic system. These systems carry fluids around the body. This means that the cancer cells can travel far from the original tumour and form new tumors when they settle and grow in a different part of the body.

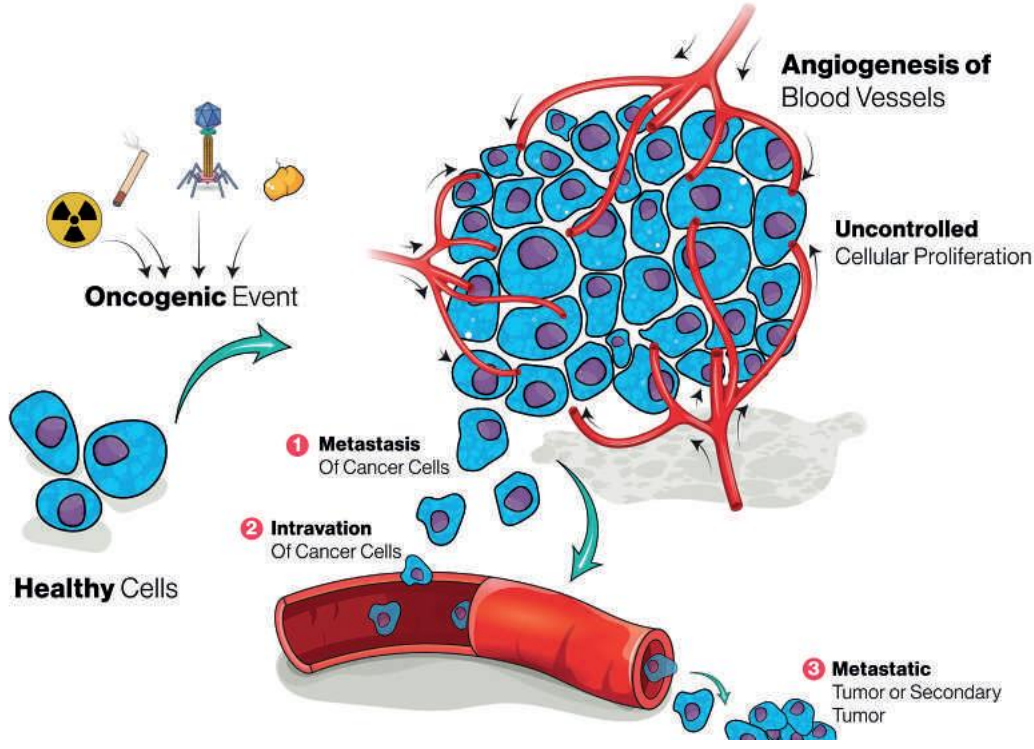


Figure 32.4: Cancer mechanism

Oncogenic event – cancer-causing event (*onco* – from Greek meaning tumour)

Oncology – study of cancers

CERVICAL SCREENING

A swab is taken from the cervix and analysed to find precancerous cervical cells.

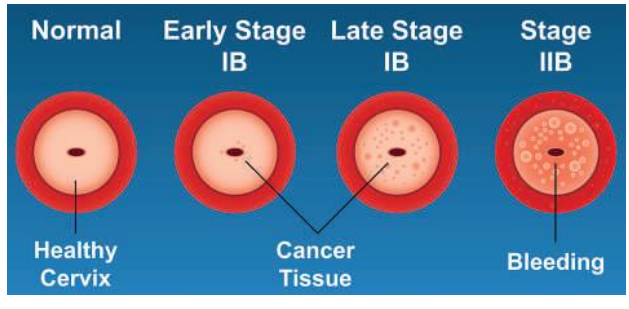


Figure 32.5: Cervical cancer stages

There are three main ways to screen for cervical cancer.

- The human papillomavirus (HPV) test checks cells for infection with high-risk HPV types that can cause cervical cancer.
- The Pap test (also called a Pap smear or cervical cytology) collects cervical cells so they can be checked for changes caused by HPV that may, if left untreated, turn into cervical cancer. It can find precancerous cells and cervical cancer cells.
- The HPV/Pap co-test uses an HPV test and Pap test together to check for both high-risk HPV and cervical cell changes.

PSA TESTING

- Prostate specific antigen is a protein made by the prostate in males.
- The PSA blood test measures the amount of prostate specific antigen in the blood.
- PSA levels are used to monitor prostate cancer.

High levels of PSA can be caused by prostate cancer, but it doesn't always mean a diagnosis of prostate cancer.

Age	Reference range
40-49	up to 2.5 ng/ml
50-59	up to 3.5 ng/ml
60-69	up to 4.5 ng/ml
70-79	up to 6.5 ng/ml

Figure 32.6: Normal levels of PSA for different age groups

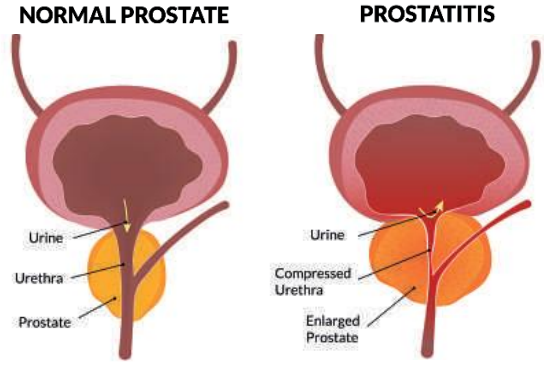


Figure 32.7: Prostate cancer

- Prostate cancer is a tumour that starts in the prostate gland.
- The prostate produces most of the fluid that makes up semen and nourishes the sperm.
- There are different types of prostate cancer. Most grow slowly and never cause harm (benign), while others spread to other parts of the body and cause serious harm, and even death (malignant).
- Prostate cancer is more common in older men. It is also more common if they have at least one close blood relative with prostate cancer (i.e. father, brother or son diagnosed under 65 years of age).

BREAST SCREENING

Breast cancer is the abnormal growth of the cells lining the breast lobules or ducts. These cells grow uncontrollably and have the potential to spread to other parts of the body. Both men and women can develop breast cancer, although it is uncommon in men.

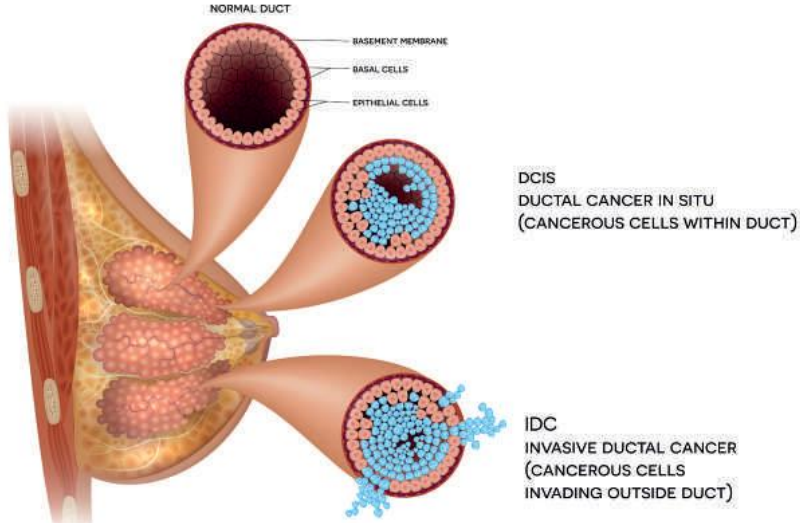


Figure 32.8: Breast cancer

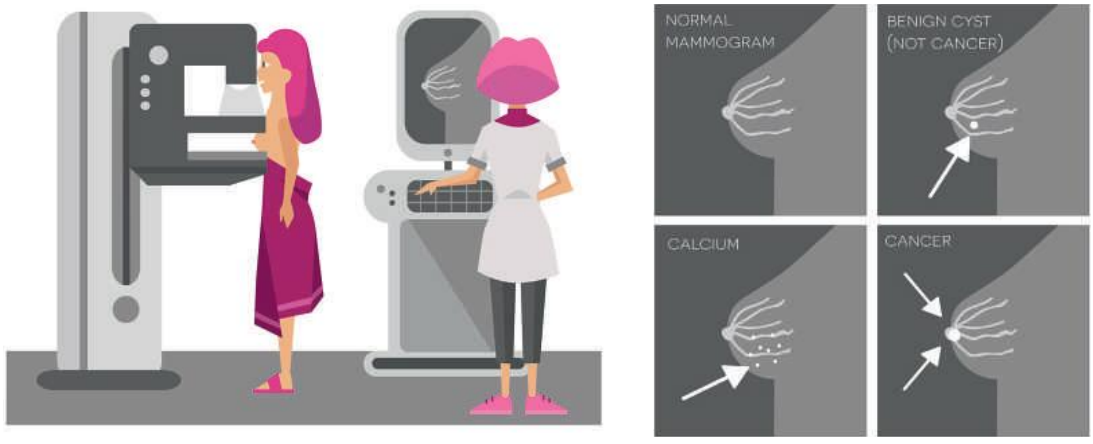


Figure 32.9: Breast screening process

During the screening procedure, the breast is squeezed vertically and horizontally to reduce the distance the X-rays need to penetrate to produce clear images. The X-ray images produced from breast screening are called mammograms.

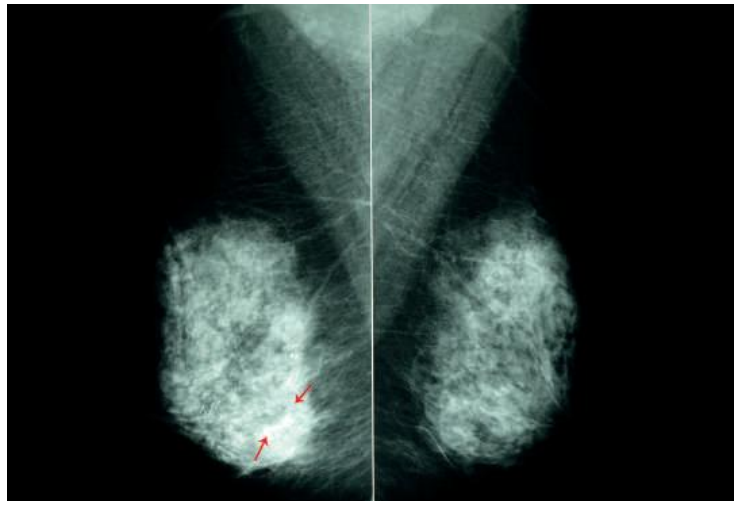


Figure 32.10: Mammogram showing cancerous (left) and normal (right) breast tissue

Some factors that increase your risk of breast cancer include:

- increasing age
- family history
- inheritance of mutations in the genes BRCA2, BRCA1 (more common with Ashkenazi Jewish heritage) and CHEK2
- exposure to female hormones (natural and administered)
- starting your period before the age of 12
- a previous breast cancer diagnosis
- a past history of certain non-cancerous breast conditions.

Lifestyle factors that can also slightly increase the risk of breast cancer in men and women include:

- being overweight
- not enough physical activity
- drinking alcohol.

There is also an association with some benign breast disease and past exposure to radiation.

Question 1

State two key qualities of stem cells. (2 marks)

Question 2

Describe the feature that distinguishes pluripotent cells from multipotent cells. (2 marks)

Question 3

State two sources of human stem cells used in medical research and treatment. (2 marks)

Question 4

Describe how cancers are caused at the cellular level. (2 marks)

Question 5

Describe the ethical problem of using embryonic stem cells. (2 marks)

Question 6

Explain why bone marrow transplants are given to people with leukemia after the patient's own bone marrow is destroyed by radiation. (3 marks)

Question 7

Suggest why cancers more common in older people. (3 marks)

Question 8

Outline how cancers can spread around the body. (5 marks)

EMBRYONIC DEVELOPMENT AND BIRTH

33

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• The development of the embryo after implantation involves the differentiation of cells into three different germ layers that will eventually produce specific systems in the body and the placenta.				
• The stages of labour include birth, during which there are circulatory system changes in the child.				

Every human started as a single cell – the zygote, formed by the fertilisation of an ovum by a sperm.

The zygote starts the divide by mitosis and by the time it travels down the fallopian tube to the uterus to implant into the endometrium it has reached the blastocyst stage.

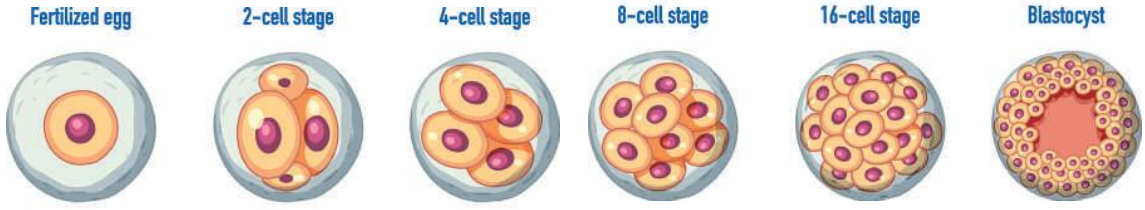


Figure 33.1: Human embryonic development

Outer cells or trophoblast becomes the amnion, chorion and placenta membranes.

Inner cell mass becomes the foetus. These cells are embryonic stem cells.

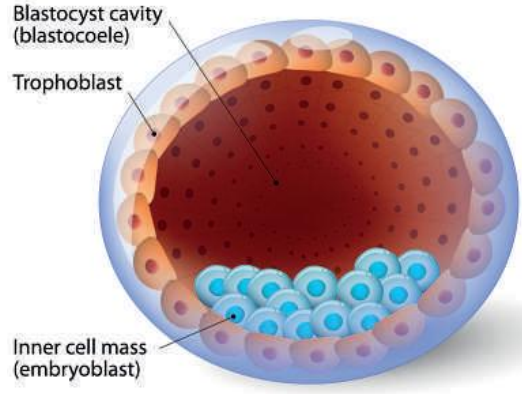


Figure 33.2: Blastocyst

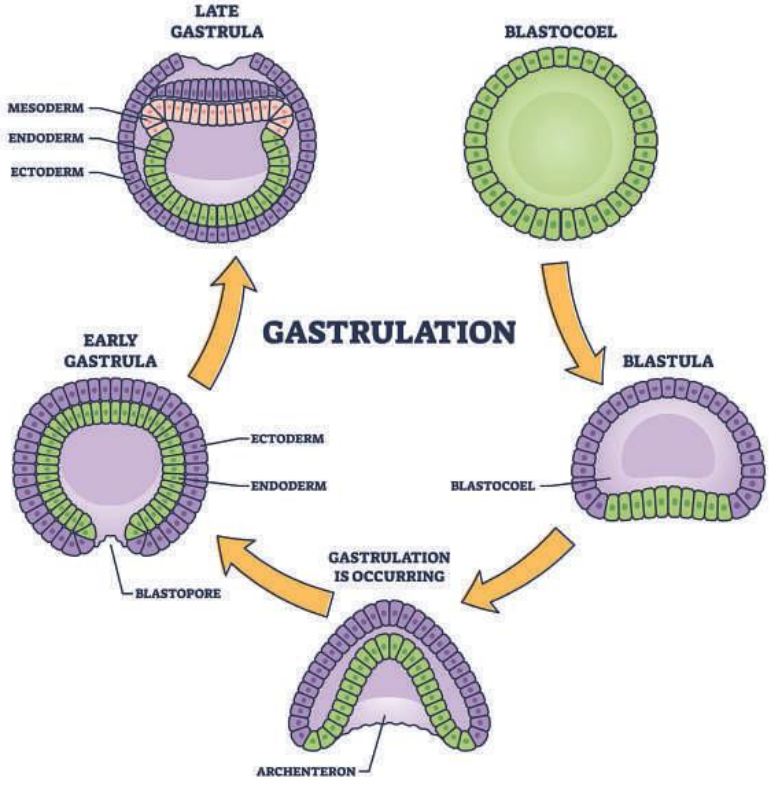


Figure 33.3: Gastrulation

The spherical ball of cells of the blastocyst, process through gastrulation to form the three main layers from which all the body cells and tissues arise. This happens late in the second week after fertilisation.

Ectoderm forms the central nervous system, skin, sweat glands, skin sensor receptors, hair follicles, the external surfaces of the eyes (cornea and lens), teeth (enamel), mouth, and rectum, as well as the pituitary glands.

Mesoderm forms the skeletal system, the muscular system, the excretory system, the circulatory system, the lymphatic system, and the reproductive system. It also gives rise to connective tissues, including blood.

Endoderm forms the digestive tract and associated organs and glands, the lungs, liver, and pancreas, thymus, thyroid, and parathyroid glands and the membranes lining the internal organs.

As the embryo develops the potency of the cells decreases, becoming more specialised to producing cells of particular tissue types, and tissues.

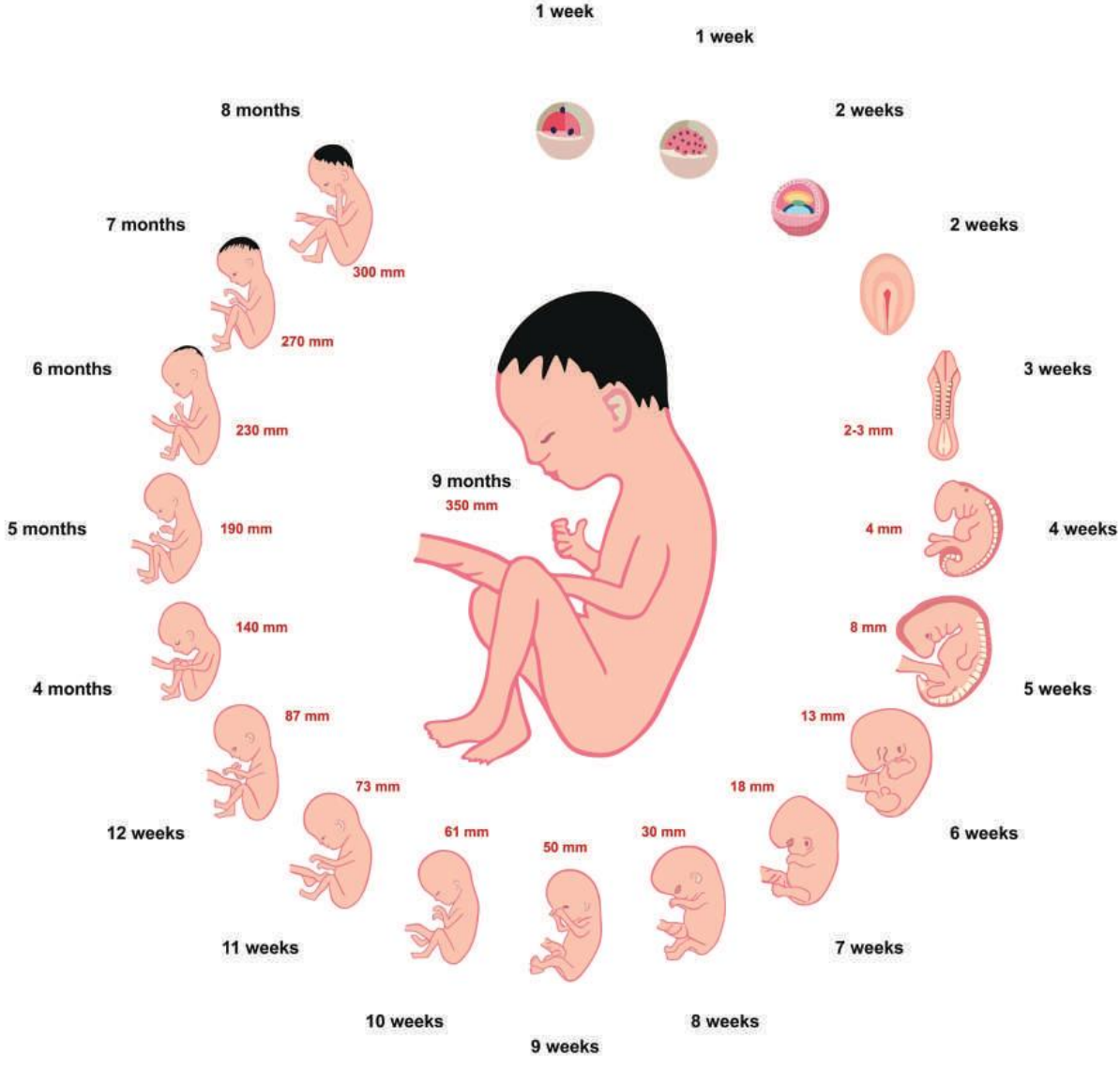


Figure 33.4: Change in form and size of the embryo and foetus during gestation

For a more in-depth outline of embryonic and foetal development go to:

https://embryology.med.unsw.edu.au/embryology/index.php/Timeline_human_development

TERATOGENS

A **teratogen** is a substance that interferes with normal foetal development and causes congenital disabilities. Drugs, alcohol, some chemicals and toxic substances are examples of teratogens. Teratogens can also increase the risk for miscarriage, preterm labour or stillbirth.

The following factors determine how dangerous teratogen exposure is during pregnancy:

- the drug, substance or type of toxin
- how long the pregnant person was exposed
- the amount of exposure (dosage or quantity)
- the gestational age of the foetus (weeks of pregnancy) at exposure
- hereditary factors that could increase the foetus's risk.

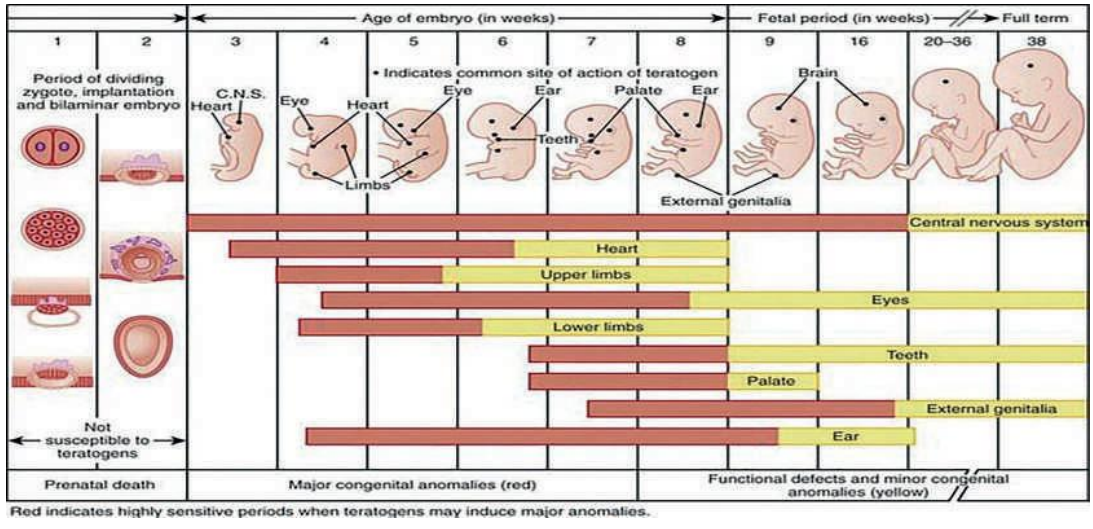


Figure 33.5: Critical periods in human development. Red indicates highly sensitive periods when teratogens may induce major anomalies. Yellow indicates minor impact of teratogens.
From: https://basicmedicalkey.com/impact-of-age-on-pharmacology/

Note the length of time that teratogens can have an impact on the central nervous system.

ALCOHOL, CIGARETTES AND RECREATIONAL DRUGS ARE KNOWN TERATOGENS

Alcohol affects the foetus's central nervous system.

- Drinking alcohol during pregnancy increases the foetus's risk for foetal alcohol syndrome. Foetal alcohol syndrome is a disorder that can cause abnormal facial features, a small head and brain and other physical and behavioural disabilities. There's no amount of alcohol intake that's considered safe during pregnancy.

Cigarette smoking is associated with foetal growth restriction, premature birth and miscarriage. Smoking also affects the foetus's sensitive lung tissue and brain. Because vapes contain very similar contents, the risks for the effects of cigarette smoking are similar to vaping.

Recreational drugs such as cocaine, methamphetamines, heroin and marijuana during pregnancy can cause low birth weight, heart problems and neonatal abstinence syndrome – when a baby goes through drug withdrawal after birth. Sharing needles can also cause infection.

Infections, viruses, parasites and other bacterial illnesses can pose serious threats to a pregnant person and the foetus, including toxoplasmosis (in cat faeces), listeria, STIs such as syphilis, hepatitis and HIV, as well as the rubella and chicken pox viruses.

Heavy metals, such as lead and mercury, and **radiation** or **chemotherapy** also affect the development of the foetus.

THE PLACENTA

The placenta develops within the uterus during pregnancy, playing a key role in providing nutrients and oxygen to the foetus, as well as removing waste materials such as urea and carbon dioxide.

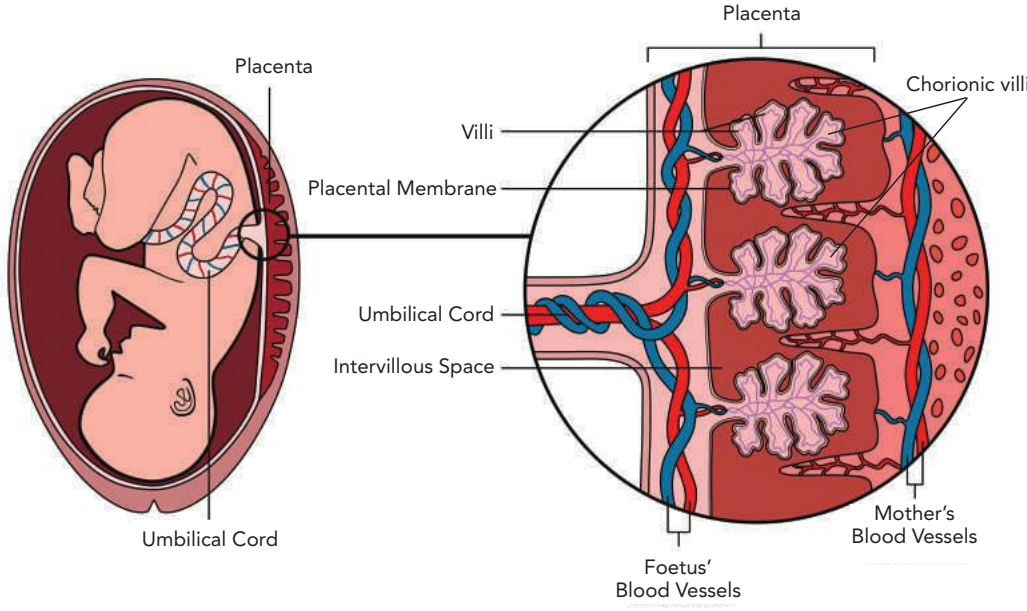


Figure 33.6: The structure of the placenta

The placental membrane forms a barrier to differentiate blood supply from the mother and the foetus, but it allows many substances to be exchanged. Note that the maternal blood fills the spaces around the chorionic villi in which the foetal blood vessels are located, providing a very large surface area to volume ratio for the exchange of materials.

The umbilical cord attaches the foetus to the placenta.

Because the placenta is the exchange surface for gases, nutrients and wastes, the foetal circulation is structured to allow for this.

- ductus arteriosus – connecting the pulmonary artery to the descending aorta. It allows most of the blood from the right ventricle to bypass the foetus's fluid-filled non-functioning lungs
- foramen ovale – hole in the septum of the heart between the left and right atria to allow blood to bypass the pulmonary circulation.

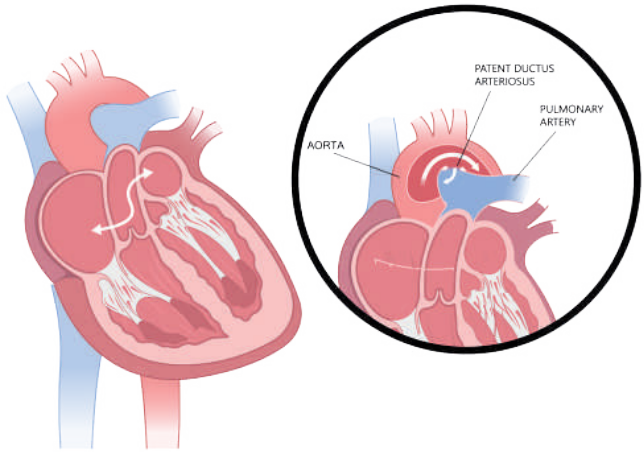


Figure 33.7: Foetal heart structures Foramen ovale (left) and Ductus arteriosus (right)

Both close soon after birth.

MULTIPLE BIRTHS

More than one fertilised ovum can be implanted into the uterus at the same time.

Monozygotic or identical or maternal twins: develop from a single fertilised ovum and formed when the cells at the 2–8 cell stages separate. Only one placenta is formed.

Dizygotic fraternal twins: two ova are released from the ovaries and fertilised by different sperm at the same time and both implant into the endometrium. Two placentas are formed.

In May 2021, woman from Mali successfully gave birth to nine babies (nonuplets) and all of them survived, making a new world record for multiple births.

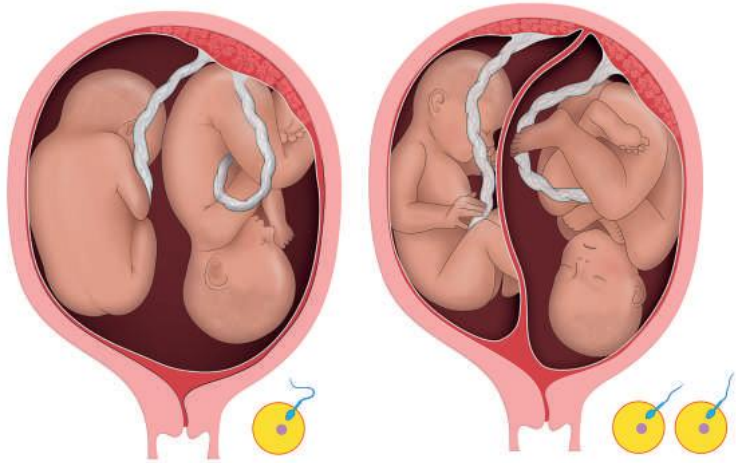


Figure 33.8: Monozygotic twins sharing a placenta and amnion (left) and dizygotic twins with separate placentas and amnions (right)

BIRTH

Oxytocin is secreted from the posterior pituitary to stimulate the uterine muscles to contract causing the onset of the birth process.

At the time of birth, the foetus normally is oriented with the head down in the pelvis area allowing the head to emerge first on delivery. This allows the baby to take its first breaths.

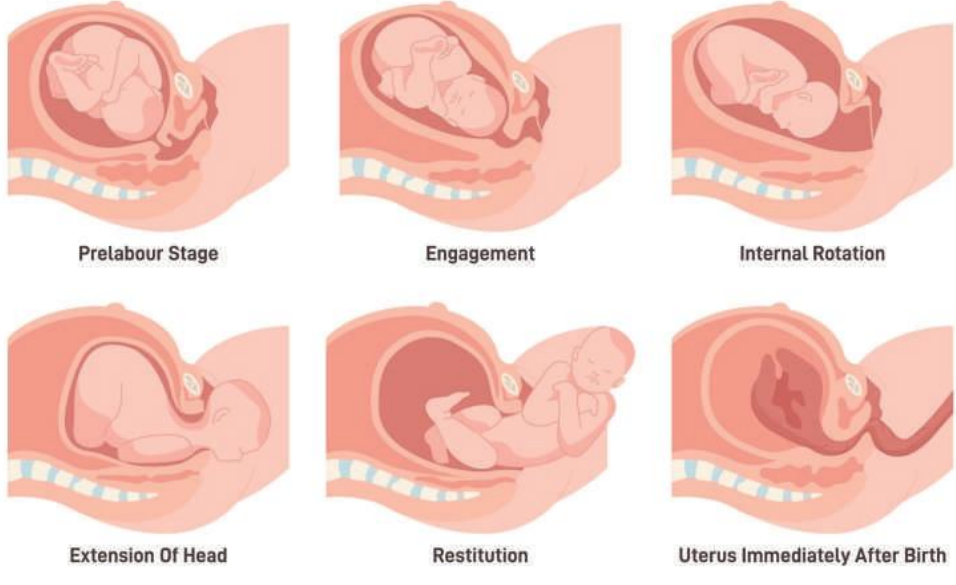


Figure 33.9: Stages of birth – vaginal delivery

FIRST STAGE

- The cervix soften and opens.
- The membranes rupture (waters break) and amniotic fluid leaks out.

SECOND STAGE

- From when the cervix is fully dilated to when the baby is born. The umbilical cord is clamped and cut.

THIRD STAGE

- The placenta is delivered.

Birth may be by Caesarean section – through an incision in the abdomen.

When the foetus is oriented with its bottom near the cervix, it is called a breech birth.

The risks of attempting a vaginal breech birth are:

- Injuries to your baby's legs or arms such as dislocated or broken bones
- The umbilical cord can be flattened or twisted during delivery. This can cause nerve or brain damage due to a lack of oxygen.

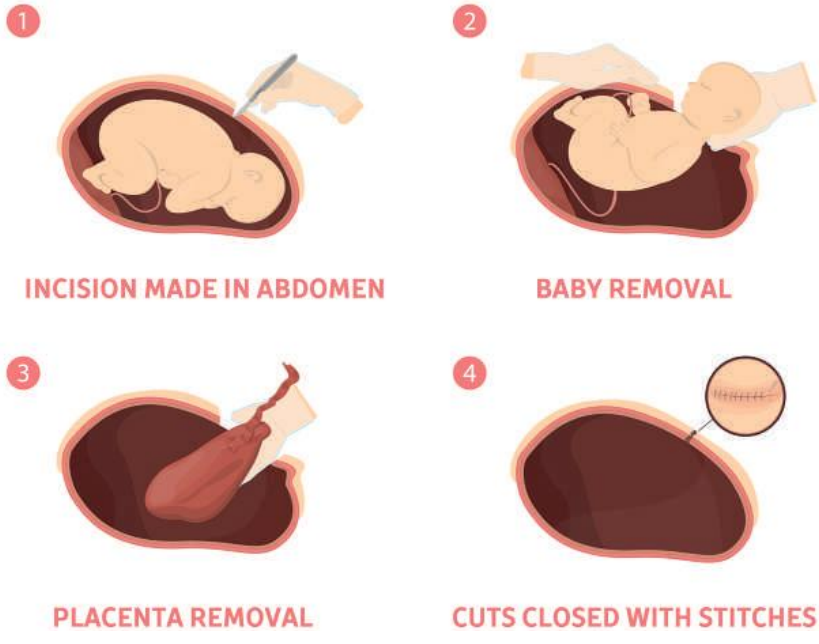


Figure 33.10: Birth via Caesarean section

Question 1

Which of the following is true for the placenta? (1 mark)

(a) It allows wastes to be eliminated from the foetus.
(b) Foetal and maternal blood mix in the chorionic villi.
(c) It transfers maternal proteins to the foetus.
(d) It provides temperature control of the foetus.

Question 2

The correct sequence for the early development following fertilisation is: (1 mark)

(a) embryo, zygote, morula, blastocyst.
(b) embryo, morula, zygote, blastocyst.
(c) morula, gastrula, blastocyst, embryo.
(d) zygote, morula, blastocyst, gastrula.

Question 3

Which of the following systems develops first during pregnancy? (1 mark)

(a) Musculoskeletal system
(b) Nervous system
(c) Digestive system
(d) Respiratory system

Question 4

Which hormone is produced by the placenta to stop the development of ovarian follicles? (1 mark)

(a) Oxytocin
(b) Oestrogen
(c) Progesterone
(d) hCG

Question 5

The lymphatic and reproductive systems of the foetus develop from which layer of the gastrula? (1 mark)

(a) Endoderm
(b) Mesoderm
(c) Ectoderm
(d) Maxiderm

Question 6

At which stage of development can the formation of identical twins or triplets occur? (1 mark)

(a) At fertilisation
(b) On implantation
(c) At the 2–8 cell stage
(d) When the morula forms

Question 7

Alcohol, cigarette smoke and radiation can have detrimental effects on foetal development. They are called: (1 mark)

(a) teratogens.
(b) carcinogens.
(c) abortogens.
(d) epimetrogens.

Question 8

The foetal circulation has the foramen ovale and the ductus arteriosus to: (1 mark)

(a) deliver blood to the placenta.
(b) bypass the non-functional foetal lungs.
(c) allow for the separation of oxygenated and deoxygenated blood.
(d) exchange more nutrients with the placenta.

Question 9

The membrane that surrounds the foetus in the uterus is called the: (1 mark)

(a) chorion.
(b) blastocoel.
(c) placental membrane.
(d) amnion.

Question 10

Which of the following is not an important factor in determining how dangerous teratogen exposure is during pregnancy? (1 mark)

(a) How long the pregnant person is exposed
(b) The week of pregnancy
(c) The age of the mother
(d) The amount of exposure

34 LIFESTYLE CHOICES

Key Teaching Points	Discover	Revise	Enrich	Exam Prep
SYLLABUS CHECKPOINT				
• Lifestyle choices, including diet, illicit drugs, alcohol and nicotine, may affect foetal development.				
• Lifestyle choices, including being active or sedentary, the use of drugs, and type of diet, can compromise body functioning in the short term and may have long-term consequences.				

We are what we eat and do

The structure and function of the systems relies on the inputs and activities of the whole body.

Balance Moderation Variety are three very important considerations in everything we do to and with our bodies.

Figure 34.1: Factors affecting fertility

Main areas in which lifestyle choices impact our wellbeing

Figure 34.2: Life balance depends on lifestyle choices

- **Diet**: Eating an unbalanced diet can have a negative impact on health.
- **Activity**: Inactivity can lead to health problems.
- **Sleep**: Poor sleep can lead to a range of health problems.
- **Alcohol use**: Drinking too much alcohol can have a negative impact on health.
- **Smoking**: Smoking, including vaping is a major cause of many health problems.
- **Use of recreational drugs**: leads to a variety of physical and mental health problems.
- **Not drinking enough water**: Dehydration can lead to health problems.

DIET

Figure 34.3: Dietary choices

Figure 34.4: https://www.eatforhealth.gov.au/guidelines/australian-guide-healthy-eating

Variety and **moderation** are key to eating a balanced diet to provide all the required energy, building materials, vitamins and minerals for a healthy body.

ACTIVITY

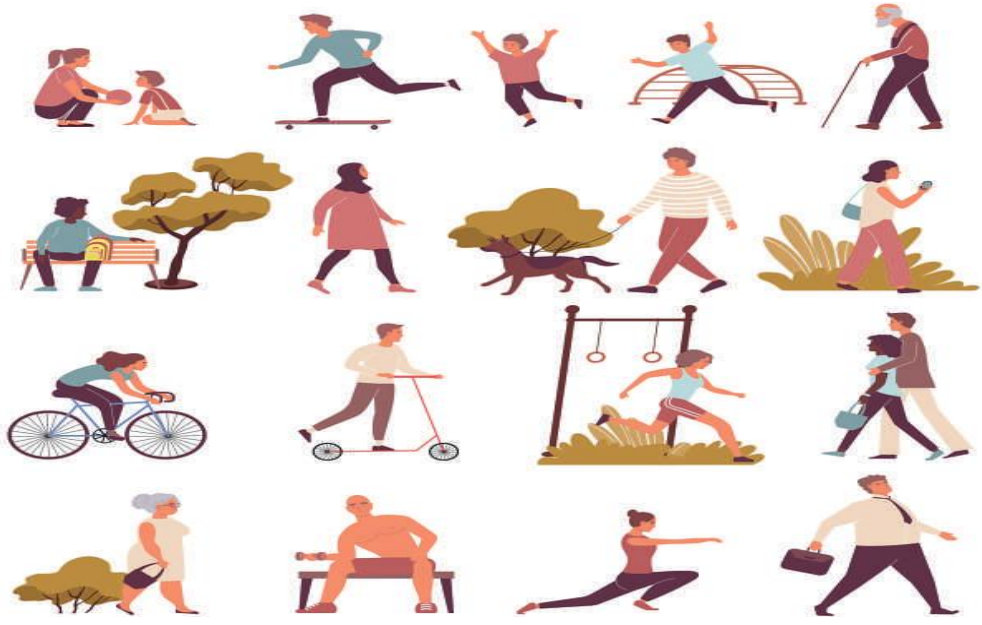


Figure 34.5: Different ways to move

One of the keys to health involves moving the body. It increases respiration and circulation, reducing the free radicals that cause damage to body tissues which can produce cancers.

SLEEP

Sleep helps keep the immune system strong and the heart and blood vessels healthy. It allows for growth and healing, and helps control appetite and weight. Sleep promotes attention, memory and learning.

Studies have shown that not getting enough quality sleep can lead to:

- higher levels of the hormones that control hunger, including leptin and ghrelin, inside your body
- decreased ability to respond to insulin
- increased consumption of food, especially fatty, sweet, and salty foods
- decreased physical activity
- metabolic syndrome raising the risk of heart disease, diabetes and stroke.

All of these contribute to overweight and obesity.

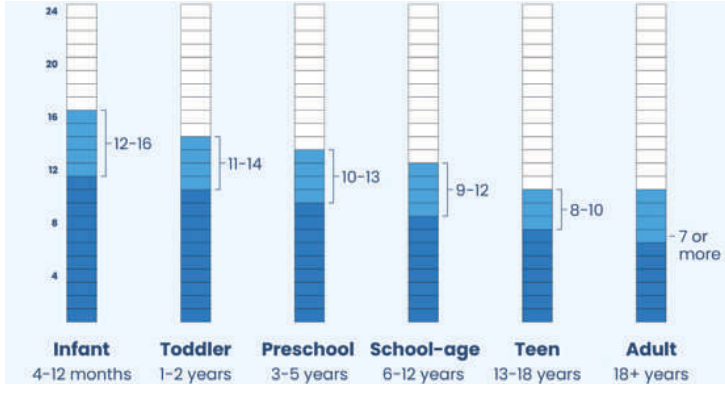


Figure 34.6: Recommended hours of sleep at different ages
From: How Much Sleep Do You Need? | Sleep Foundation – Sourced from American Academy of Sleep Medicine

ALCOHOL USE

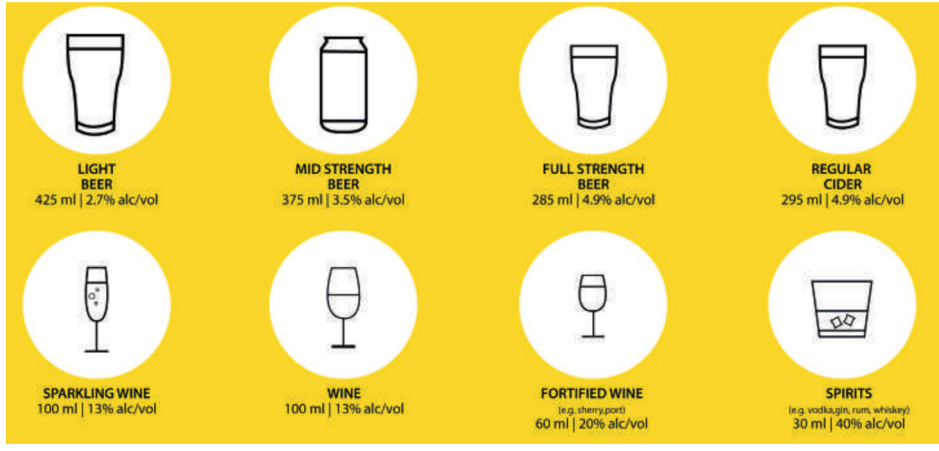


Figure 34.7: Standard drink sizes for different alcoholic drinks

These guidelines are from https://www.health.gov.au/news/australian-alcohol-guidelines-revised

Following the guidelines keeps the risk of harm from alcohol low, but it does not remove all risk. Healthy adults drinking within the guideline recommendations have less than a 1 in 100 chance of dying from an alcohol-related condition.

Guideline 1: Reducing the risk of alcohol-related harm for adults

To reduce the risk of harm from alcohol-related disease or injury, healthy men and women should drink no more than 10 standard drinks a week and no more than 4 standard drinks on any one day. The less you drink, the lower your risk of harm from alcohol.

Guideline 2: Children and people under 18 years of age

To reduce the risk of injury and other harms to health, children and people under 18 years of age should not drink alcohol.

Guideline 3: Women who are pregnant or breastfeeding

To prevent harm from alcohol to their unborn child, women who are pregnant or planning a pregnancy should not drink alcohol. For women who are breastfeeding, not drinking any alcohol is safest for their baby.

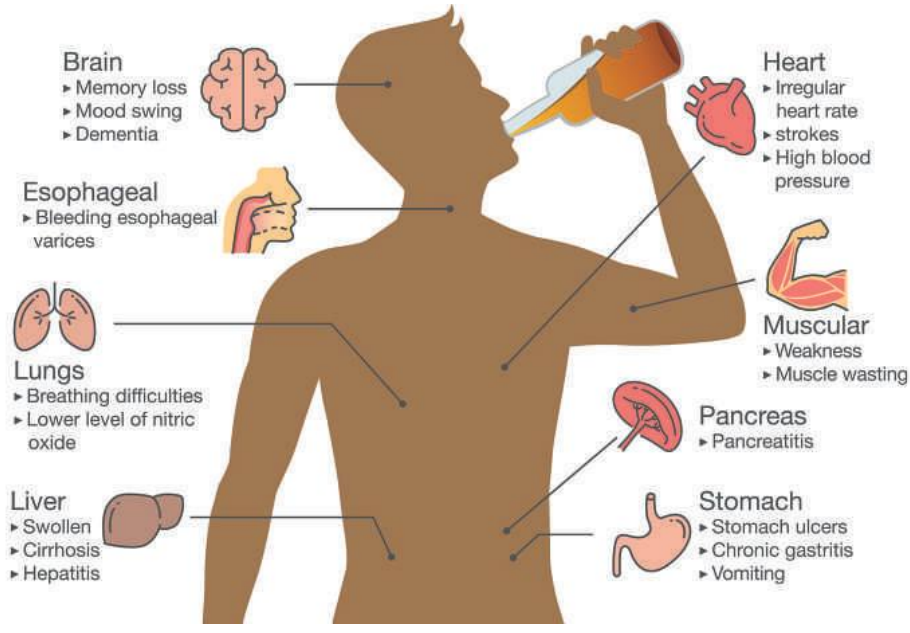


Figure 34.8: Symptoms and effects of alcoholism

SMOKING

Smoking has many long-term adverse effects on the body.

- It reduces sperm count.
- It increases the risk of pregnancy loss and congenital disabilities.
- It increases the risk of cataracts.
- It impairs immune system function.
- It increases general inflammation.
- It can cause cancer in nearly any part of the body, including the lungs, kidneys, and stomach.
- It triggers asthma attacks.
- It causes blockages in the veins and arteries.
- It increases the risk of a stroke.

The long-term health effects of vaping found that people using e-cigarettes had a higher risk of respiratory disease than people who never smoked.

Vaping may:

- damage the lungs
- release free radicals into the body, which promote cancer development
- weaken the immune system
- delay brain development in foetuses, children, and teenagers.

For non-smokers, the comparison should be between vaping and breathing air. Vaping is clearly more harmful than breathing air.

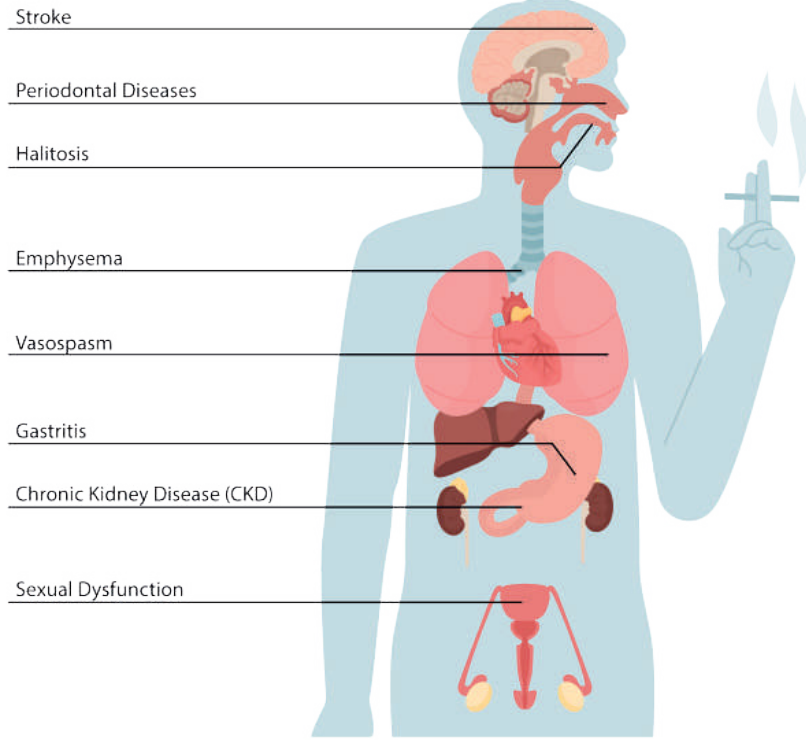


Figure 34.9: Effects of smoking on the body

RECREATIONAL DRUGS

CANNABIS

Prior to 2016, cannabis was classified as an illegal narcotic under Australian law. This changed in February 2016, when the *National Drugs Amendment Act 2016* established a national licensing and permit scheme to enable the cultivation, production, and manufacture of cannabis for medicinal and related research purposes. Under this scheme, specific patient groups can now access medicinal cannabis products under strict medical supervision. Cannabis cultivated for other purposes remains illegal.

Short-term effects	Long-term effects
• Mild euphoria, relaxation and reduced inhibitions • Perceptual alterations, including time distortion and intensification of ordinary experiences • Feelings of hunger • Panic reactions, confusion and feelings of paranoia – mainly reported by naïve users • Nausea, headache and reddened eyes • Increased heart rate for up to 3 hours after smoking • Dizziness, with impaired balance and coordination	• Physical dependence • Upper respiratory tract cancers, chronic bronchitis and permanent damage to the airways when smoked • Cardiovascular system damage • Mental health conditions including depression • Poor adolescent psychosocial development

Source: Adapted from (Hall & Degenhardt 2009; Nielsen & Gisev 2017; NSW Ministry of Health 2017).

Figure 34.10: Effects of cannabis
From: https://www.aihw.gov.au/reports/alcohol/alcohol-tobacco-other-drugs-australia/contents/drug-types/cannabis

METHAMPHETAMINES

Also called chalk, crystal, glass, ice, meth and speed.

Short-Term Effects

Taking even small amounts of methamphetamine can result in many of the same health effects as those of other stimulants, such as cocaine or other amphetamines. These include:

- increased wakefulness and physical activity
- decreased appetite
- faster breathing
- rapid and/or irregular heartbeat
- increased blood pressure and body temperature.

Long-Term Effects

Methamphetamine use has many other negative consequences, including:

- extreme weight loss
- addiction
- severe dental problems
- intense itching, leading to skin sores from scratching
- anxiety
- changes in brain structure and function
- confusion
- memory loss
- sleeping problems
- violent behaviour
- paranoia – extreme and unreasonable distrust of others
- hallucinations – sensations and images that seem real though they aren't.

ECSTASY OR MDMA

People who use MDMA regularly can experience long term effects, including:

- problems with memory and concentration
- depression
- high blood pressure
- cracked teeth from clenching and grinding
- liver problems.

COCAINE

People who use cocaine regularly experience:

- poor mental function
- poor sexual performance
- bronchitis
- anxiety
- high blood pressure
- paranoia
- seizures.

Task 34.1: Mark your level of risk on the following scales.

Diet
Balanced ---------- Unbalanced

Activity
Inactive ---------- Overactive

Sleep
Recommended level ---------- Poor sleep

Alcohol use
None ---------- Too much

Smoking
Non-smoker ---------- Heavy smoker

Recreational drugs
Non-user ---------- Heavy user

Water intake
Too little ---------- Too much

Comment on your overall health risk according to the information in your scales.

TASK SUGGESTED SOLUTIONS

UNIT 1

CHAPTER 1: CELLS AND BODY HIERARCHY

Task 1

A car is not living.

– it does not reproduce
– do not grow through internal processes

Features that could be considered as living

– it has a highly organised structure
– responds to 'stimuli' – pressure on accelerator or brake
– requires a source of energy to function
– models change over time e.g. petrol cars to hybrids to electric cars
– requires inputs – fuel, oil, water, produces outputs – exhaust gases
– life span – indeterminate

To be considered living, it needs to satisfy **all** the features.

Task 2

System	Organs
Musculoskeletal system	bones, muscles
Circulatory system	heart, arteries, veins, capillaries lymph vessels, lymph nodes bone marrow, thymus. spleen, tonsils
Respiratory system	nose, pharynx, trachea, bronchi,, lungs, diaphragm
Nervous system	brain, spinal cord, nerves, sensory organs – eyes, ears, tongue
Endocrine system	hypothalamus. pituitary gland, thyroid gland, ovaries, testes, pancreas
Digestive system	teeth, salivary glands, pharynx, oesophagus, stomach, small intestine, large intestine, liver, gall bladder, pancreas
Urinary or excretory system	kidney, bladder, ureters, urethra, liver
Reproductive system	ovaries, uterus, fallopian tubes, testes, penis
Integumentary system	skin, hair, nails, sweat glands sebaceous glands, breasts

Task 3

Lings – respiratory
Heart – circulatory
Brain – nervous
Liver – digestive, excretory
Thyroid – endocrine
Nasopharynx – respiratory, digestive
Male reproductive system – reproductive, endocrine
Female reproductive system – reproductive, endocrine
Intestines – digestive
Kidneys – excretory
Bladder – excretory
Thymus – immune
Muscle – musculoskeletal

Artery – circulatory
Blood – circulatory
Skin – integumentary
Pancreas – endocrine, digestive
Spleen – circulatory, immune
Gall bladder – digestive
Knee joint – musculoskeletal

CHAPTER 2: CELLULAR STRUCTURE

Task 1

All body cells are different, but have common features.
The generalised cell shows all the common features found in the variety of body cells.

CHAPTER 3: TISSUE TYPES AND STEM CELLS

Task 1

Smooth muscle tissue – regular, constant contractions (peristalsis) churn food in the stomach and move it into the small intestine
Loose connective tissue – supports the inner most layer of the stomach and the blood vessels, lymph vessels and nerves supplying the stomach
Nervous tissue – controls the movement of muscles and opening and closing of the cardiac and pyloric sphincters.
Blood – absorbs nutrients from the stomach; supply the tissues of the stomach with oxygen and nutrients and removes wastes and carbon dioxide
Columnar epithelium – produces mucus that protects the stomach lining from enzyme action; produces acid and pepsinogen (precursor to the enzyme pepsin)

Task 2

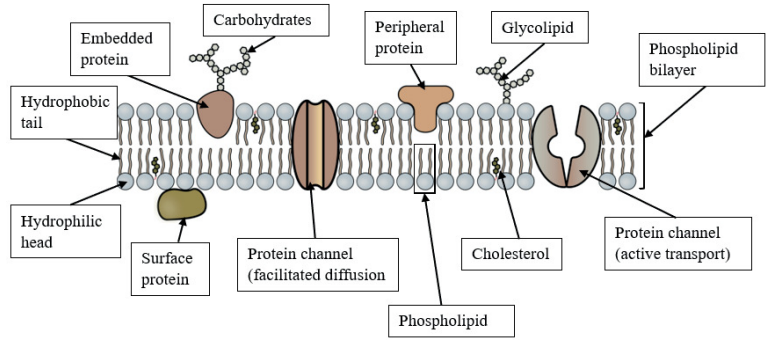
Simple squamous – lining blood vessels, alveoli
Simple cuboidal – lining the nephrons of the kidney and ducts of glands
Simple columnar – lining digestive tract and upper respiratory tract
Stratified squamous – lining of the mouth and vagina

CHAPTER 4: CELL MEMBRANE AND THE EXCHANGE OF MATERIALS

Task 1

Lower surface is the internal surface.
Evidence

- glycolipids and glycoproteins (carbohydrate/embedded protein combination) only occur on the outside of cells as they act as receptors for environmental change
- surface proteins are found inside the cell



Task 2

	Diffusion	Facilitated diffusion	Osmosis	Active transport
Down a concentration gradient	yes	yes	yes – the concentration of water in a solution	no
Against a concentration gradient	no	no	yes – when considering the concentration of the solutes in a solution	yes
Energy requirement	none	none	none	yes
Substances moved	oxygen, carbon dioxide	ions and small polar molecules e.g. glucose, sodium ions and chloride ions	water	materials needed to be moved from an area of low concentration to an area of higher concentration; materials that need to be moved in large quantities
Notes		uses carrier proteins	requires a semi-permeable membrane to separate to solutions	can use carrier proteins or the cell membrane engulfs materials. Vesicles from within the cell merge with the cell membrane to release materials to the outside

CHAPTER 5: METABOLISM

Task 1

Respiration is the only reaction in the body that uses oxygen and produces carbon dioxide.

The breakdown of glucose uses a known amount of oxygen to produce a known amount of energy (about 16 kJ/g of glucose) and a known amount of carbon dioxide. Measurement of oxygen use or carbon dioxide production will be an indirect measure of the amount of energy released during respiration for use in cells. This is the ONLY source of energy for cellular function – metabolism.

Task 2

The overall reaction of the breakdown of glucose on the barbecue releases a large amount of energy in one step. This also requires large amounts of energy to start the reaction and the large amounts of energy released will heat the surroundings to very high temperatures.

Cells breakdown glucose in small steps to control the amount of energy released at any one time to maintain the optimum temperature conditions for the functioning of the cell, and releasing energy in amounts that can be readily used to produce ATP from ADP.

Task 3

Anaerobic respiration in cells would occur when the cells are not supplied with sufficient amounts of oxygen to maintain aerobic respiration ie. respiration is using oxygen faster than it can be supplied by the blood. Skeletal muscles can go into anaerobic mode when athletes push their limits.

Task 4

For a person who weighs 60 kg and is 1.55m tall.

BMI = body weight (kg)/height (m)^2

e.g. BMI = $60/(1.55)^2$ = $60/2.4$

BMI = 25

Task 5

Increased exercise or activity reduces the risks of developing obesity when comparing two people with the same dietary intake. Exercise/activity requires energy. If it is not used the energy-carrying molecules (glucose) will be stored as fats → obesity.

CHAPTER 6: FOOD TYPES AND DIETS

Task 1

Macronutrient	Sources
Proteins	meat, eggs, dairy products, potatoes, nuts and seeds,
Carbohydrates	fruit, cereals, nuts, potatoes, sweets/lollies, bread/cakes
Lipids	oils, nuts and seeds, avocadoes, olives, eggs, fish, meat, dairy products

Task 2

Uses of lipids in the body

- long term energy storage in the form of fats
- production of cellular components such as cell membranes
- help in the production of hormones
- protection of nerve cells and internal organs
- help in the absorption of fat soluble vitamins

Task 3

Vitamin	Sources in the diet
A	dairy products, oily fish, eggs, carrots, squash, sweet potato, kale, spinach
B	meat, eggs, seafood, dairy products, legumes, leafy greens, seeds
C	citrus fruit, capsicum, tomatoes, strawberries, broccoli, cauliflower
D	mushrooms, oily fish, egg yolks, dairy products, almonds
Iron	meat, fish, eggs, nuts, leafy green vegetables
Calcium	dairy products, soy products, green leafy vegetables, almonds
Magnesium	spinach, legumes (beans and peas), nuts and seeds, whole grains
Iodine	fish and other seafood, eggs, dairy products, iodized table salt
Zinc	seafood, meat, pumpkin seeds, oats, milk
Selenium	meat, fish, Brazil nuts
Sodium	table salt, cheese, cured meats e.g. ham, packaged crisps

Task 4

Iron intake

Omnivorous diet – meat, seafood, eggs, cereals, leafy greens e.g. spinach

Vegan diet – nuts and seeds, leafy greens e.g. spinach, soy beans, cereals

Haem iron sources found in meats are easily used by the body.

Non-haem iron is harder to absorb and is found in plant-based foods.

CHAPTER 8: ENZYMES

Task 1

Enzymes reduce the activation energy required for a reaction to occur. Each of the steps in the cellular respiration reaction require low activation energy which means they can occur at body temperature.

CHAPTER 10: CIRCULATORY SYSTEM

Task 1

Depends on personal circumstances.

CHAPTER 11: BLOOD AND TRANSFUSIONS

Task 1

S – AB+

T – B+

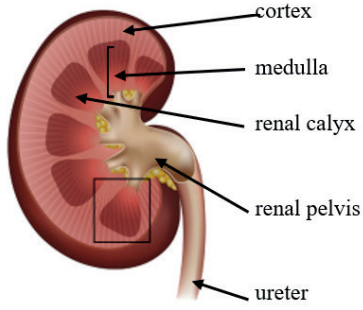
Task 2

Universal recipient – AB+ – blood cells have all antigens so will not react to any in the transfused blood

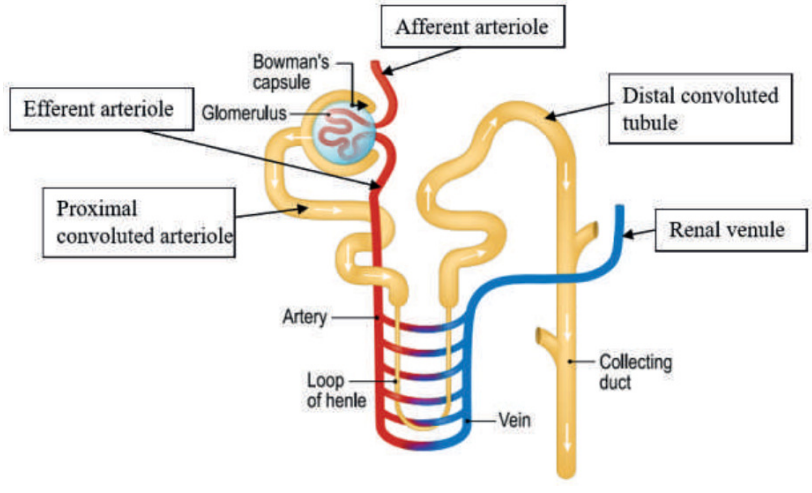
Universal donor – O- – blood cells contain no antigens to cause a reaction in recipients

CHAPTER 14: EXCRETORY SYSTEM

Task 1



Task 2



	Function
A	bring blood to the glomerulus
B	takes blood from the glomerulus to the loop of Henle
C	capillary bed that provides a large surface area over which filtration of the blood can occur
D	takes filtered materials from the blood into the nephron
E	reabsorbs glucose, amino acids and much of the water from the filtrate
F	concentrates the filtrate by removing water
G	active secretes materials into the filtrate
H	takes the urine from the nephron to the pelvis of the kidney
I	takes deoxygenated blood from the nephron back into the general circulation

CHAPTER 15: SKELETON

Task 1

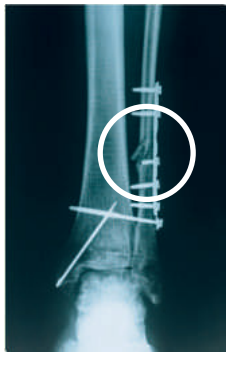
Axial skeleton	Appendicular skeleton	
cranium	pectoral girdle and arms	pelvic girdle and legs
jaw	clavicle	ilium
ear ossicles	scapula	ischium
vertebrae	humerus	pubis
ribs	ulna	femur
sternum	radius	patella
sacrum	carpals	tibia
	metacarpals	fibula
	phalanges	tarsals
		metatarsals
		phalanges

CHAPTER 16: JOINTS

Task 1

Movement at joints	Type of movement
A	flexion
B	rotation
C	flexion
D	flexion
E	rotation
F	flexion
G	abduction, adduction
H	abduction adduction
I	circumduction
J	abduction
K	flexion
L	abduction
M	extension
N	supination
O	dorsiflexion
P	dorsiflexion
Q	plantar flexion

Task 2



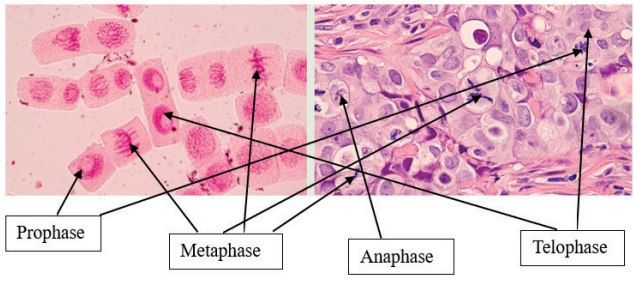
UNIT 2

CHAPTER 18: MITOSIS AND MEIOSIS

Task 1

Stage of mitosis	Cellular activities
Interphase	DNA replicates; growth of the cell; production of cell organelles
Prophase	chromosomes condense and become visible; spindle fibres form and attach to centromeres of chromosomes; nuclear membrane breaks down
Metaphase	chromosomes move to align along the equator of the cell moved by the spindle fibres
Anaphase	centromere divides; spindle fibres contract drawing the chromatids apart moving them to opposite poles of the cell
Telophase	spindle fibres detach; chromosomes dissipate so are no longer visible; nuclear membranes form around the two sets of chromosomes
Cytokinesis	division of the cytoplasm; distribution of organelles to the two cells; cell membrane moves to cause a constriction between the two nuclei; cell membrane fuses to form two separate cells

Task 2



CHAPTER 19: DNA

Task 1

DNA sequence: GAT AAA TCT GGT CTT ATT TCC

RNA sequence: CUA UUU AGA CCA GAA UAA AGG

Amino acid sequence: Leu – Phe – Arg – Pro – Glu – STOP – Arg

Task 2

Culprit: Suspect 2 – has the same lines on the electropherogram as the material from the crime scene.

Suspect 2 could have shed DNA at the scene, prior to the incident; or Suspect 2's DNA could have been deposited on the clothing of the victim prior to the incident; Suspect 2 could have sneezed in the area where the victim was found.

Task 3

Need to match the STR profiles to see which ones are the same: Yes, the profile of the suspect was the same as the sample taken from the crime scene and different to that of the victim.

CHAPTER 21: EPIGENETICS

Task 1

Epigenetic factors – work on the genome and change how the genome is expressed

Methylation – methyl groups attach to the DNA at certain locations; stop transcription from happening

Epigenetic markers can be on the histone proteins

Acetylation – acetyl groups attached to certain parts of the histones; determines how tightly the DNA is wrapped around the histones; acetyl present → loose packing around histone allows for transcription of DNA; acetyl absent → tight packing of DNA no access for transcription

Epigenetic markers impact which genes are expressed.

Environment and diet impact expression of genes

Epigenetic markers are inherited through mitosis

Epigenetic marks can be 'cleared' at fertilization, some are not.

Internal cues (epigenetic factors) work for human development and functions of specific cells/tissues.

In cancer, the epigenetic markers are often not arranged as they should be.

CHAPTER 24: PEDIGREE CHARTS

Task 1

Genotype	Phenotype (blood group)
$I^a I^a$	A
$I^a I^b$	AB
$I^b I^b$	B
$I^a i$	A
$I^b i$	B
ii	O

CHAPTER 25: MALE REPRODUCTIVE SYSTEM

Task 1

Testosterone regulates	Secondary sex characteristics
sex drive	development of testes and penis
bone mass	increase in size/height
fat distribution	at deposits in abdomen, chest and shoulders
muscle mass	increase in muscle mass (effects athletic ability)
production of red blood cells	increased production of red blood cells (effects athletic ability)
production of sperm	essential for producing sperm at a rate that determines fertility; too much reduces sperm production

Task 2

Head defects

- effects the acromere and the amount of enzyme required to penetrate the corona radiata
- double head too much DNA and embryo would not survive

Mid-piece defects

- bent, asymmetric – won't move efficiently through the female reproductive tract
- errors in the number of mitochondria would reduce the energy supply for movement of the sperm

Tail defects

- all effect the way in which the sperm moves and the speed of movement

CHAPTER 28: SEXUALLY TRANSMITTED INFECTIONS

Task 1

Types of contraception	**Effectiveness (%)** providing the methods are used correctly and consistently. Percentage decreases dramatically if method is not use properly.
Abstinence	100
Withdrawal	78
Douche	depends on contents of douche; but low effectiveness overall
Rhythm method	76-88

Types of contraception	**Effectiveness (%)** providing the methods are used correctly and consistently. Percentage decreases dramatically if method is not use properly.
Condoms	85
Diaphragm	88
Contraceptive pill	91
Contraceptive injection	94
Contraceptive implant/patch	99
Intrauterine device (IUD)	99
'Morning after' pill	85
RU-486 pill	96
Sterilization	99
Termination/abortion	100

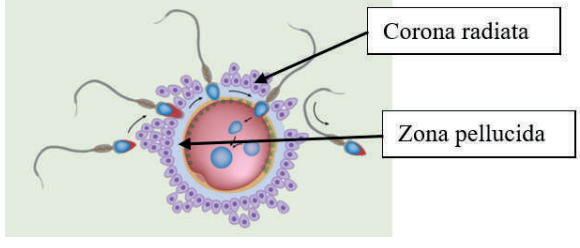
Task 2

Factors affecting effectiveness of contraceptive methods

- timing of use
- consistency of use
- contents of treatment
- correct use of method

CHAPTER 29: CONCEPTION AND IMPLANTATION

Task 1

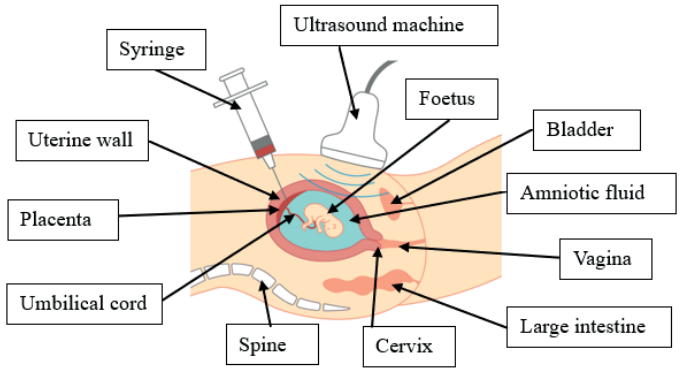


CHAPTER 31: GENETIC TESTING AND EMBRYONIC SCREENING

Task 1

Figure 2: 2 Y chromosomes
Figure 3: 1 sex chromosome (X only)
Figure 4: 3 sex chromosomes (2 Xs)
Figure 5: 3 chromosome 18
Figure 6: 3 chromosome 21
Figure 7: addition to one of chromosome 14

Task 2



ANSWERS TO REVISION QUESTIONS

UNIT 1

CHAPTER 1: CELLS AND BODY HIERARCHY

Question 1

Description	Marks
Respiratory system	1
Circulatory system	1
Total	**2**

Question 2

Description	Marks
(a) muscular, skeletal, nervous systems	3
(b) reproductive system	1
(c) endocrine system	1
(d) respiratory and digestive systems (pharynx); circulatory and immune systems (white blood cells); male urinary and reproductive systems (urethra)	3
(e) reproductive system	1
Total	**9**

Question 3

Description	Marks
Hair is the product of cells and the remains of dead cells and has no functioning cellular contents	1
bone is made up of living cells with a hard matrix between the cells	1
Total	**2**

Question 4

Description	Marks
The dead cells are the remains of once living cells.	1
For these to accumulate at the base of the nail, living cells need to have died and be replaced by other living cells,	1
which is an indication of growth.	1
Total	**3**

Question 5

Description	Marks
An unconscious person will still respire	1
therefore there will be gas exchange and movements of the respiratory system as well as movements within the circulatory system such as a heartbeat	2
In death, gas exchange ceases and no circulation of blood occurs.	2
An unconscious person may also show reflexes but a dead person will not	2
Total	**7**

Question 6

Description	Marks
Decomposition	1
Total	**1**

Question 7

Description	Marks
Respiratory system Excretory system Digestive system Reproductive system	4
Total	**4**

Question 8

Description	Marks
Tissues are made up of similar cells with similar functions	2
Organs are made up of different tissues that carry out different functions but work together	2
Total	**4**

Question 9

Description	Marks
Fatigue, weakness Pale skin Cold hands and feet Shortness of breath	4
All due to a lack of oxygen getting to cells for respiration to supply energy	2
Total	**6**

Question 10

Description	Marks
Lack of enzymes would cause very little digestion of the food as the breakdown without enzymes occurs at a very low rate	2
Lack of acid would result in conditions not appropriate for the functioning of enzymes	2
Total	**4**

Question 11

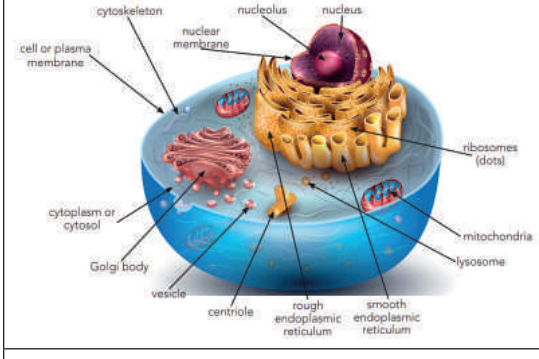
Description	Marks
Nutrients can be given intravenously i.e. put directly into circulation	1
In forms that can be used by cells	1
Total	**2**

Question 12

Description	Marks
Muscles are required to move air into and out of the lungs for gas exchange and for the beating of the heart.	2
If this doesn't occur, then the person will suffocate	1
Total	**3**

CHAPTER 2: CELLULAR STRUCTURE

Question 1

Description	Marks
	14
Total	**14**

Question 2

Description	Marks
(a) B, E, G	3
(b) A – yellow one; D – central big one or one tiny outside; F – one strand of the three shown	3
(c) A – blue dot; D – blue dots in small cells, pink in central cell (upper left and lower right) E – pink oval; F – blue dots	4
(d) E is has the ability to move using its own energy using its flagellum	1
(e) B and C have cellular extensions	1
(f) Both require a lot of energy to move or contract – mitochondria supply energy in the form of ATP	2
(g) G could not grow, reproduce/undergo mitosis, repair itself	3
(h) Ribosomes, endoplasmic reticulum and Golgi bodies – all involved in producing protein secretions	4
Total	**21**

CHAPTER 3: TISSUE TYPES AND STEM CELLS

Question 1

Description	Marks
(a) epithelium (stratified)	1
(b) epithelium (ciliated columnar)	1
(c) epithelium (cuboidal)	1
(d) epithelium (squamous)	1
(e) nervous	1
(f) reproductive (ovum)	1
(g) connective (adipose)	1
(h) reproductive (sperm)	1
(i) connective (bone)	1
(j) muscle (smooth	1
(k) muscle (striated/voluntary)	1
(l) connective (blood)	1
(m) muscle (smooth/involuntary)	1
Total	**13**

Question 2

Description	Marks
The contents or flexibility of the matrix surrounding the cells	1
Total	**1**

Question 3

Description	Marks
Epithelial tissue	1
Total	**1**

Question 4

Description	Marks
The presence of striations	1
Voluntary or involuntary control	1
The presence of branching structures between the cells	1
Total	**3**

Question 5

Description	Marks
Cuboidal epithelium	1
Total	**1**

Question 6

Description	Marks
Schwann cells – produce the myelin sheath around axons;	2
glial cells – produce support for the structure and function of nerve cells	2
Total	**4**

Question 7

Description	Marks
Cilia help move materials across the surface of the tissue.	1
They are found in the trachea and fallopian tubes	1
Total	**2**

Question 8

Description	Marks
Smooth muscle tissue automatically and constantly applies varying pressure to vessels and organs, causing movement of materials in the area.	2
Striated muscle would not be suitable because it would need constant thought to maintain the movement.	2
Total	**4**

Question 9

Description	Marks
Blood is a connective tissue because it has cells in a matrix (plasma)	1
Total	**1**

Question 10

Description	Marks
A tissue is made up of similar cells with similar functions	2
An organ is made of several different types of tissue that work together to bring about a function	2
Total	**4**

Question 11

Description	Marks
Pluripotent cells can differentiate into any body tissue cells w	1
Multipotent stems cells can only make cells of the same tissue category	1
Total	**2**

Question 12

Description	Marks
Embryonic stem cells are taken from an embryo and are naturally pluripotent	2
Adult stem cells are found in areas of the body where new cells are formed and they are multipotent	2
Total	**4**

Question 13

Description	Marks
Stem cells form the new cells that replace cells that are damaged or lost during the injury	2
Total	**2**

CHAPTER 4: CELL MEMBRANE AND THE EXCHANGE OF MATERIALS

Question 1

Description	Marks
(a) Materials are able to move into and out of the cell solid the solid line does not allow for this. Microscopic examination of cells in the 1700s showed that the cell had a membrane and the level of magnification and clarity of images suggested a solid line type membrane.	3
(b) A solid membrane with gaps allowed for the flow of materials across the membrane, but not for active transport. There had to be something else in the membrane that helped control the movement of materials.	3
(c) The solid line with 'gated gaps' did not allow for the movement of materials that were either water soluble or lipid soluble and some materials that were too large to move across by themselves.	3
(d) The bilayer of phospholipids conformed with the more up to date micrographs from electron microscopes. 'Gates' or different protein masses could also be seen.	2
(e) Chemical analysis and scanning electron microscopy identified the attached protein and carbohydrate chains and the movement of the protein channels across the membrane.	3
Total	**14**

Question 2

Description	Marks
Molecules are always in motion.	1
In nature, molecules tend to move randomly to areas where they are less concentrated	1
Due to the collisions between like molecules forcing them away from one another.	1
Total	**3**

Question 3

Description	Marks
The more the moving molecules bump into one another the further they spread out.	2
Temperature increases the speed of movement of the molecules, making the collisions between them more dramatic and so the molecules move further with each collision.	2
Diffusion or movement of the molecules from where they are concentrated to where they are less concentrated occurs quicker.	2
Total	**6**

Question 4

Description	Marks
The semipermeable membrane allows only some molecules to pass across from one side to the other by diffusion (small molecules) or by osmosis (water)	2
Other molecules are retained within the cell or restricted from entering the cell unless aided by the inclusions in the cell membrane and the use of energy.	2
The cell can control the materials that enter and leave the cell to maintain an optimum internal environment in a changing external environment	2
Total	**6**

Question 5

Description	Marks
Large particles such as viruses or fragments of destroyed cells or	1
large amounts of fluids containing materials that can't cross the membrane	1
Total	**2**

Question 6

Description	Marks
Exocytosis	1
Total	**1**

CHAPTER 5: METABOLISM

Question 1

Description	Marks
The inside of the mitochondria has a large surface area of membranes.	1
The enzymes required to catalyse the reactions in aerobic respirations are located on the membranes.	1
The greater the surface area, the greater the number of enzymes and so the greater the rate at which the reactions occur	1
Total	**3**

Question 2

Description	Marks
(a) growing adolescents – have a higher metabolic rate because they require larger amounts of energy to provide for the growth and development of the body; adults need to only maintain the body that has grown to its maximum	2
(b) current netball players – require a lot of energy to train and play at maximum effort so their metabolic rate is high; retired netball players don't exercise to the level of the current players so their energy needs are less	2
(c) pregnant women have a higher metabolic rate to provide for the growth of the embryo and placenta and the greater functioning of the maternal organs such as the kidney due to the presence of the foetus; non-pregnant women do not have the extra mass to develop or maintain	2
Total	**6**

Question 3

Description	Marks
As glycogen in muscles and the liver	1
As lipids in adipose tissue	1
Total	**2**

Question 4

Description	Marks
The liver has a very high level of metabolic activity	1
Temperature relies on the waste energy released during the chemical reactions of metabolism	1
Total	**2**

Question 5

Description	Marks
Blood supplies oxygen for respiration in cells.	1
There is no change in the metabolic activity of the brain during exercise compared to normal waking activity, so no change in energy required	2
Muscles require a great increase in the amount of oxygen supplied to the cells to keep up with the rate of oxygen supply for use in cellular respiration to produce energy for the increase in the amount of movement.	2
Total	**5**

Question 6

Description	Marks
Metabolic rate increases when the body is exposed to very high temperatures because the body undertakes energy-requiring activities to actively lose heat eg. sweating.	2
Metabolic rate at very low temperatures increases to keep up with the energy release as heat in comparison to that which is lost from the body	2
Total	**4**

CHAPTER 6: FOOD TYPES AND DIETS

Question 1

Description	Marks
Macronutrients – required in large amounts in the diet	1
Micronutrients – required in very small amounts in the diet	1
Total	**2**

Question 2

Description	Marks
Left and right	2
Both are outside the limits set by the dietary guidelines and internationally accepted BMI range for healthy weight	2
Total	**4**

Question 3

Description	Marks
protein – Biuret test - purple	3
sugar – Benedict's test - green to orange-red	3
starch – Iodine test – blue-black	3
lipids – Brown paper test – translucent patch	3
lipids – Ethanol test - cloudy	3
Total	**15**

Question 4

Description	Marks
Nitrogen is found only in proteins not carbohydrates or lipids	2
Total	**2**

Question 5

Description	Marks
Protein is required to build physical structures of cells and tissues	1
produce new cells that increase the body mass	1
Total	**2**

Question 6

Description	Marks
The outer covering of fruit and vegetables contain high amounts of fibre compared with the rest of the fruit.	1
It may also contain the supply of vitamins attributed to that fruit or vegetable	1
Total	**2**

Question 7

Description	Marks
Hard tack and salted meat do not supply all the vitamins and minerals for the body to function properly.	2
The addition of limes or other fruit or vegetables provided a source of vitamins and minerals that reduced the risk of deficiency diseases.	2
Total	**4**

Question 8

Description	Marks
Eating a whole fresh orange or apple provides a good source of fibre and the sugar and vitamin contents of the fruit.	2
A glass of juice may contain the juice from 3 or 4 fruits which will provide higher levels of sugars but not the fibre. Will provide excess amount of vitamins that will be excreted.	2
The fibre will help the person feel fuller and limit further intake of food, therefore having a impact on weight control.	2
Total	**6**

Question 9

Description	Marks
These diets will have different sources of the same nutrients	1
in different ratios	1
Total	**2**

Question 10

Description	Marks
There are different dietary recommendations for different groups of people because each group has different requirements for growth and maintaining metabolism.	1
The guidelines are given to maintain a healthy weight and metabolism.	1
Total	**2**

CHAPTER 7: DIGESTIVE SYSTEM

Question 1

Description	Marks
mouth → oesophagus → stomach → small intestine → large intestine	5
Total	**5**

Question 2

Description	Marks
Lips Epiglottis Cardiac Sphincter Pyloric Sphincter Ileo-Caecal Sphincter Anus	6
Total	**6**

Question 3

Description	Marks
The food decays	1
Causes inflammation of the appendix	1
If the appendix is not removed, it can burst and cause inflammation and infection of the peritoneal cavity and death	2
Total	**4**

Question 4

Description	Marks
Rumbling is caused by peristalsis	1
Moving gases in the digestive tract.	1
Total	**2**

Question 5

Description	Marks
Bacteria break down materials such as fibre that human digestion can't.	2
The products of this breakdown can be absorbed and used by the body	2
Total	**4**

Question 6

Description	Marks
Stomach banding reduces the volume of the stomach so it is full with less food.	2
The food is not stored in the smaller stomach so will move through the stomach faster.	2
Therefore less food will pass though the digestive tract more quickly	2
Total	**6**

Question 7

Description	Marks
Mechanical digestion increases the surface area on which enzymes can work	2
Enzymes and chemical reactions only occur on surfaces	1
Total	**3**

Question 8

Description	Marks
Stomach contents are acidic	1
When acid enters the oesophagus, it irritates the lining	1
The oesophagus is not protected by as much mucus as is the stomach	1
Discomfort and pain will result	1
Total	**4**

Question 9

Description	Marks
The digestive tract is very long, allowing time for digestion to occur	2
The folded nature of the linings of the tract provide for a greater surface area through which nutrients can be absorbed	2
Total	**4**

Question 10

Description	Marks
A shorter large intestine will impact the ability to absorb nutrients and water from the contents.	2
If the lower end of the large intestine is removed, then the formation of faeces will be adversely affected	2
Total	**4**

CHAPTER 8: ENZYMES

Question 1

Description	Marks
Optimum conditions are those that allow the enzyme to work at its maximum rate	1
Total	**1**

Question 2

Description	Marks
An active site is the location on the enzyme molecule where the substrate attaches to cause a reaction to occur	1
Total	**1**

Question 3

Description	Marks
• reaction occurs at a lower temperature	1
• reaction requires less energy to occur	1
• energy is released in amounts that are useful to produce ATP from ADP and not heat the surrounding to excessive levels	1
Total	**3**

Question 4

Description	Marks
Co-enzymes are non-protein molecule that bind to the enzyme	1
Completing the shape of the enzyme	1
To bring about the attachment of the substrate to the active site to bring about a reaction	1
Total	**3**

Question 5

Description	Marks
The lack of this enzyme stops the cascade of reactions required to cause blood clotting	1
Therefore clotting does not occur and the wound continues to bleed	1
Total	**2**

Question 6

Description	Marks
The kinetic theory of matter states that molecules move faster at higher temperatures.	1
This will cause the molecules in the reaction to collide at higher speeds	1
Therefore there is energy for the reaction to take place faster.	1
Enzymes are protein based	1
At higher temperatures will cause the molecular structure to breakdown	1
Changing its shape so it no longer fits the active site on the substrate to bring about a reaction	1
Total	**6**

Question 7

Description	Marks
Substances of different pH can change the rate of action of enzymes e.g. acid action on pepsin in the stomach	2
The alkali present in the duodenum reduces pepsin activity but allows for trypsin to work.	2
Some substances can attach to the substrate blocking the active site reducing reaction rates.	2
Other substances can help in the binding of enzymes to the active site	2
Other substances can help in the transfer of energy in the reaction chain increasing reaction rates	2
Total	**10**

Question 8

Description	Marks
Different sections of the digestive tract have different pH allowing for different enzymes to act.	2
Total	**2**

Question 9

Description	Marks
There are three or more steps required to breakdown proteins to their basic units of amino acids.	1
Each protein and its reactions require different enzymes.	1
Proteins have quaternary structures that have to be broken down to smaller proteins, then large protein chains, then smaller protein chains then, dipeptides, then amino acids – many steps each requiring different enzymes	3
Lipases have only one step in their digestion	1
Total	**6**

Question 10

Description	Marks
Bile emulsifies the fat globules producing smaller spheres with greater surface-area-to-volume ratio	2
This allows lipases to work on increased surface area, increasing the rate of the reaction	2
Total	**4**

CHAPTER 9: LIVER

Question 1

Description	Marks
storage of glucose as glycogen	1
breakdown of glycogen to glucose	1
produces cholesterol or allows increased storage	1
breaks down alcoho	1
breaks down toxins	1
storage as proteins or deamination of excess amino acids	1
storage of triglycerides as fat	1
breaks down penicillin	1
breaks down hormones	1
excretes it into the bile as bile salts	1
stores vitamins or breaks down excess	1
stores iron and copper	1
breaks down to produce bile salts	1
activates by changing structure	1
breaks down hormones to produce hydrogen carbonate	1
excretes to bile	1
Total	**16**

CHAPTER 10: CIRCULATORY SYSTEM

Question 1

Description		Marks
Moving out of the blood at capillaries	Moving into the blood at capillaries	
oxygen	carbon dioxide	2
nutrients	wastes	2
hormones		1
heat	heat (depending on temperature difference)	2
water	water	2
	Total	**9**

Question 2

Description	Marks
The heart is a double pump separating oxygenated and deoxygenated blood.	2
This means the cells will be supplied with the greatest concentration of oxygen available and not diluted by blood from the pulmonary circulation.	2
For an efficient supply of oxygen to active cells	1
Total	**5**

Question 3

Description	Marks
The hole between the ventricles allows blood from the pulmonary and systemic circulatory paths to mix	2
Reducing the level of oxygen supplied to active cells which in turn impacts on the rate of cellular respiration.	2
Low rates of cellular respiration effects metabolism, growth and levels of activity in the child	2
Total	**6**

Question 4

Description	Marks
A heart murmur can indicate that blood is not flowing efficiently through the valves of the heart.	1
This could allow for the backflow of blood between the ventricles and atrium	1
Which would mean inefficient circulation of blood carrying nutrients and gases to places of exchange	1
Total	**3**

Question 5

Description	Marks
The aorta is the blood vessel that is subjected to the greatest amount of pressure from the beating on the heart.	2
It has to be able to expand and contract to allow for the blood to pass and not rupture	2
Total	**4**

Question 6

Description	Marks
The heart muscle tissue needs to be supplied with nutrients and gas the same as any other tissue.	1
The blood in the heart chambers is separated from the cardiac tissues cells by thick, impermeable membranes.	1
The heart has its own set of blood vessels called coronary blood vessels	1
That branch into capillaries that are very close to the cardiac muscle cells to allow for the exchange of materials	1
Total	**4**

Question 7

Description	Marks
The clot blocks the flow of blood to the capillary bed,	1
There can be no exchange of materials from the blood to the cells.	1
Once the cells have used their supply of oxygen they die	1
Total	**3**

CHAPTER 11: BLOOD AND TRANSFUSIONS

Question 1

Description	Marks
(a) agglutination	1
(b) O-	1
(c) Rhesus factors (Rh)	1
(d) haemoglobin	1
(e) leucocytes	1
Total	**5**

Question 2

Description	Marks
Erythrocytes have no nucleus so cannot repair themselves	2
As they get older they become more rigid and are damaged	1
To an extent that they are inefficient at carrying oxygen and are removed form circulation	2
Total	**5**

Question 3

Description	Marks
Oxygen can be carried attached to haemoglobin the erythrocytes	1
or dissolved in the plasma	1
Total	**2**

Question 4

Description	Marks
Iron is required to make haemoglobin.	1
Lack of iron means less haemoglobin is produced	1
Therefore less oxygen can be carried	1
Total	**3**

Question 5

Description	Marks
Different leucocytes have specific functions in the immune responses	2
Total	**2**

Question 6

Description	Marks
As blood passes through areas of high metabolic rate, such as the liver, it is heated by conduction.	1
As it flows through areas of lower metabolic activity and where the temperature is lower, the heat energy moves out of the blood into the cooler surrounding tissues.	1
Total	**2**

Question 7

Description	Marks
Haemophiliacs can be given plasma because it contains the clotting factor they are lacking	2
Total	**2**

Question 8

Description	Marks
The blood removed during a donation needs to be replaced.	1
It takes that long for the extra blood cells to be produced on top of those that needed to be replaced normally	2
Total	**3**

Question 9

Description	Marks
Arteries carry oxygenated blood.	1
When oxygen combines with haemoglobin, blood becomes bright red.	1
When oxygen is removed from the haemoglobin, it changes to a dark red colour.	1
Veins are close to the skin surface and when seen, they appear blue in colour	1
Total	**4**

CHAPTER 12: LYMPHATIC SYSTEM

Question 1

Description	Marks
Tonsilitis is inflammation of the tonsils	1
caused by an increase number of lymphocytes and phagocytes needed to engulf and remove the pathogens that have come into the mouth and invaded tissues of that area	2
Total	**3**

Question 2

Description	Marks
The movement of lymph through the lymph vessels has been blocked	1
The fluid accumulates in the muscle and other tissues of the leg	1
Total	**2**

Question 3

Description		Marks
Plasma	Lymph	
Contains no cells	Contains lymphocytes, phagocytes and other types of white blood cells	2
Flows within blood vessels	Flows within lymph vessels	2
Contains relatively high amounts of proteins for the coagulation of blood	Has very low concentrations of blood coagulating proteins	2
Fats present in plasma come from sources other than the intestines	Takes part in fat absorption form the intestine	2
Transport of dissolved nutrients and gases	Not involved in the transport of nutrients or gases	2
	Total	**10**

Question 4

Description	Marks
The lymph nodes are removed to get rid of any cancer-causing cells that may have been drained from the tissue surrounding the cancer and have moved to the nodes..	2
These may leak from the nodes and cause another cancerous nodule elsewhere in the body.	2
Total	**4**

Question 5

Description	Marks
Phagocytes engulf damaged cells or their fragments as well as pathogens including bacteria and viruses	1
That have been collected in the lymph from surrounding tissues.	1
Total	**2**

CHAPTER 13: RESPIRATORY SYSTEM

Question 1

Description	Marks
Nostril → nasal cavity → pharynx → larynx → trachea → bronchi → bronchioles → alveoli	8
Total	**8**

Question 2

Description	Marks
During a cough, the ribcage moves further in and down	1
The diaphragm contracts more and quicker than normal	1
Reducing the volume of the lungs	1
Producing a greater difference between the air pressure inside and outside of the lungs.	1
This causes the air to move out with greater force than a normal breath	1
Total	**5**

Question 3

Description	Marks
During a cold, mucus build up in the nasal cavity	1
Causes the person to breathe through their mouth.	1
The dust particles and bacteria in the air are not removed	1
Move further into the respiratory system	1
Before being captured which could cause further infection	1
Total	**5**

Question 4

Description	Marks
The epiglottis does not close over the entrance of the trachea quickly enough to stop food entering the trachea.	1
This stops the movement of air into and out of the lungs	1
A sudden increase in the pressure in the lungs caused by a hard pat on the back or the Heimlich manoeuvre (sudden strong pressure to the abdomen between the navel and ribcage)	1
Could dislodge the material blocking the trachea	1
Total	**4**

Question 5

Description	Marks
When swallowing the trachea is closed by the epiglottis.	1
The larynx is the top section of the trachea and needs to have air moving across the vocal cords to make sounds.	1
This can't happen when the trachea is closed	1
Total	**3**

Question 6

Description	Marks
The tidal volume will increase.	1
The person will breathe deeper and exhale more deeply,	1
Moving more air into and out of the lungs with each breath	1
This is done to keep up with the gas exchange requirements during exercise	1
Total	**4**

Question 7

Description	Marks
Erythrocytes contain haemoglobin to which oxygen attaches	1
Oxygen is taken out of solution in the plasma.	1
This reduces the concentration of oxygen in the plasma.	1
This maintains the high concentration difference between the blood and the air in the alveoli,	1
Promoting rapid diffusion of oxygen into the blood	1
Total	**5**

Question 8

Description	Marks
The diaphragm can still move up and down	1
Changing the volume and pressure within the lungs	1
Which causes the movements of air into and out of the lungs	1
Total	**3**

Question 9

Description	Marks
Air flow is required at a constant effective rate to remove carbon dioxide from the blood	1
and to supply the blood with oxygen.	1
Oxygen is taken to cells for energy production by respiration.	1
If the rate of oxygen supply is reduced, then energy levels decrease	1
due to lower than normal respiration rates	1
Total	**5**

Question 10

Description	Marks
Emphysema – reduces the surface of the alveoli over which gas exchanges occurs by breaking down the walls of the alveoli	2
Pneumonia – fluid fills the alveoli reducing the free surface across which oxygen can diffuse into the blood and increasing the thickness through which the oxygen needs to diffuse to reach the blood	2
Vaping and asthma – reduce the flow of air into and out of the lungs by reducing the diameter of the airways. The movement of air into and out of the lungs reduces the efficiency of gas exchange at the alveoli	2
Total	**6**

CHAPTER 14: EXCRETORY SYSTEM

Question 1

Description	Marks
(a) Filtration – between the glomerulus and Bowman's capsule	1
(b) Active reabsorption – proximal and distal convoluted tubules	1
(c) Active secretion – distal convoluted tubule	1
(d) Concentration of urine – loop of Henle, distal convoluted tubule	1
Total	**4**

Question 2

Description	Marks
kidney malfunction	1
inflammation/infection cause membranes of the glomerulus to allow larger molecules through to the filtrate	1
the amount of reabsorption is compromised because the membranes are affected by the infection/inflammation	1
glucose in urine could indicate diabetes, which causes there to be high levels of glucose in blood	1
Total	**4**

Question 3

Description	Marks
Warm, moist areas are perfect for bacterial and fungal growth.	1
The nitrogenous wastes in urine would be useful for the growth of specific micro-organisms	1
Total	**2**

Question 4

Description	Marks
Pressure causes urine to collect in the kidney rather than drain away to the bladder.	1
This affects the efficiency of the nephron function..	1
It could also cause infectious bacteria to travel up the ureter to the kidney	1
Total	**3**

Question 5

Description	Marks
Short distance for micro-organisms to travel to cause infection	1
keep area clean and dry to discourage the growth of microorganisms	1
wipe from front to back to prevent faecal matter from entering urethra and causing infection	1
Total	**3**

Question 6

Description	Marks
(a) increased volume decreased concentration	1
(b) decreased volume increased concentration	1
(c) decreased volume increased concentration	1
(d) increased volume decreased concentration	1
Total	**4**

Question 7

Description	Marks
60 kg body weight, 6 L (60kg x 100mL = 6000mL = 6L) of blood	1
180/6 = 30	1
therefore blood is filtered 30 times per day	1
Total	**3**

CHAPTER 15: SKELETON

Question 1

Description	Marks
A. scapula	1
B. humerus	1
C. ulna	1
D. metatarsals	1
E. phalanges	1
F. calcaneus (tarsals)	1
G. fibula	1
H. tibia	1
I. patella	1
J. pelvis	1
K. metacarpals	1
L. carpals	1
M. radius	1
N. lumbar vertebrae	1
O. sternum	1
P. rib	1
Q. clavicle	1
R. mandible	1
S. cranium	1
Total	19

Question 2

Description	Marks
Can live without appendicular skeleton.	1
Humans can live without moving but not without protection or support for the internal organs and central nervous system	2
Total	**3**

Question 3

Description	Marks
Look for osteons – the circular patterns of lamellae around Haversian canals produced by osteocytes in compact bone	1
If not present, it is spongy bone	1
Total	**2**

Question 4

Description	Marks
Pectoral girdle is attached to the spine by tendons and ligaments holding the scapula and clavicle in place.	2
Pelvic girdle is made up of the ilium, ischium and pubis bones attached by slightly moveable or fibrous joints to the sacrum which if part of the spinal column	2
Total	**4**

Question 5

Description		Marks
Arm	**Leg**	
humerus	femur	2
	patella	1
ulna	tibia	2
radius	fibula	2
carpals	tarsals	2
metacarpals	metatarsals	2
phalanges	phalanges	2
	Total	**13**

Question 6

Description	Marks
Hand • opposable thumb • for manipulation of objects	2
Foot • big toe in line with other toes • has two arches formed by the shape and joints between the bones • for weight-bearing in upright stance	3
Total	**5**

Question 7

Description	Marks
Absorbs force to the upper limb by transferring it to the axial skeleton, particularly the ribcage	1
Suspends the scapula (shoulder blade) so that the upper limb is able to move freely for its full range of motion	1
Protects the nerves and vessels of the upper limb that pass through the neck-armpit canal and the apex of the lung	1
Total	**3**

Question 8

Description	Marks
Ossicles are the three little bones found in the middle ear	1
Total	**1**

Question 9

Description	Marks
Reduces friction between bones	1
in moveable joints.	1
Total	**2**

CHAPTER 16: JOINTS

Question 1

Description	Marks
Joints are surrounded by ligaments, tendons and muscles that are attached to bones restricting the movement of bones in the joints.	1
The shape of the bones in the joint also determines the extent of movement..	1
Total	**2**

Question 2

Description	Marks
Level of activity	1
Amount of weight-bearing exercises	1
Smoking	1
Alcohol intake	1
Adequate intake of calcium and vitamin D is also important to maintain bone density	1
Total	**5**

Question 3

Description				Marks
elbow	hinge	ulna humerus	Flexion, extension	3
ankle	gliding	tarsals	Surface moving over another without any rotary or angular motion.	3
wrist	gliding	carpals	Surface moving over another without any rotary or angular motion.	3
base of thumb	saddle	phalanges metacarpals	Flexion, extension, adduction, abduction, and circumduction but no axial rotation.	3
neck	pivot	atlas axis	One bone rotates around another – rotation of head	3
			Total	**15**

Question 4

Description			Marks
Feature (1)	Hip (2)	Shoulder (2)	
The bones forming the joint	ilium, ischium, pubis – all fused together to form the solid socket of the acetabulum for the head of the femur	Scapula – acromium and coracoids processes protect the glenoid cavity which forms a shallow socket for the head of the humerus	5
The strength of the joint	Secured by a strong fibrous joint capsule and a number of powerful muscles; has many ligaments attaching the head of the femur to the acetabulum	Has three ligaments holding the bones together, but most of the strength comes from the muscles and tendons that cross over the joint	5
The range of movements both are capable of: extension, flexion, abduction, adduction, rotation, circumduction	Bears the weight of the body so is stronger and more stable than the shoulder; has the same range of movements but no to the same extent	Has the largest range of movements of any synovial joint in the body; stability has been sacrificed for mobility	5
		Total	**15**

Question 5

Description			Marks
Immoveable joints (1)	Between the bones of the cranium (sutures)	To form a solid inflexible bony casing for protection of the brain	3
	Between the teeth and the jaw bones	To anchor the teeth firmly so they don't fall out	2
Synovial joints (1)	Hinge joint between the mandible (lower jaw) and the cranium	Allows for chewing motions	3
	Joints between the ossicles in the middle ear	To allow for the transfer of sound waves to the inner ear	2
	Pivot join between the cranium and the vertebrae	Allows for nodding of the head – flexion and extension	2
		Total	**12**

Question 6

Description	Marks
The functions of a bursa are to facilitate movement and reduce friction between moving parts	1
Helps the muscles and tendons slide freely as the knee moves	1
Total	**2**

CHAPTER 17: MUSCLES

Question 1

Description	Marks
(a) extension	1
(b) abduction	1
(c) supination	1
(d) plantar flexion	1
Total	**4**

Question 2

Description	Marks
Tendons are made of non-elastic fibrous connective tissue.	1
This allows them to join to the periosteum of the bone and the muscle fibres producing a non-stretching connection between the two.	1
Total	**2**

Question 3

Description	Marks
(a) gastrocnemius and soleus	1
(b) hamstrings	1
(c) trapezius	1
(d) deltoid	1
(e) gluteals	1
(f) triceps	1
Total	**6**

Question 4

Description	Marks
Muscles controlling the fingers are located in the lower arm/forearm.	1
The muscles are attached by tendons to the phalanges at one end and the ulna and radius at the other	1
Total	**2**

Question 5

Description	Marks
The actin and myosin fibres are locked into position not allowing any sliding movement, so the muscles appear stiff and immoveable	2
Total	**2**

Question 6

Description	Marks
The muscle has to have one stationary end	1
and one moveable end to bring about movement.	1
Total	**2**

UNIT 2

CHAPTER 18: MITOSIS AND MEIOSIS

Question 1

Description	Marks
Anaphase	1
Total	**1**

Question 2

Description	Marks
Interphase	1
Total	**1**

Question 3

Description	Marks
Centromere	1
Total	**1**

Question 4

Description	Marks
Prophase 1	1
Total	**1**

Question 5

Description	Marks
Each of the two daughter cell in mitosis requires the same number of chromosomes as the parent cell	2
Each of the four daughter cell in meiosis requires the full N number of chromosomes	2
Total	**4**

Question 6

Description	Marks
Meiosis produces haploid cells because at fertilisation, two haploid cells (gametes with N number of chromosomes) combine to form the zygote with the correct diploid number (2N number of chromosomes).	2
Total	**2**

Question 7

Description	Marks
Interphase, when the DNA replication takes place just before prophase.	2
Metaphase, when the homologous chromosomes can cross over.	2
Anaphase, when the chromosomes may not separate completely	2
Total	**6**

Question 8

Description	Marks
Differences could arise due to errors in DNA replication;	1
swapping of chromosome sections during crossing over events to produce differences;	1
each set of parental chromosomes that make up the diploid number contain different alleles for the same gene	1
Total	**3**

Question 9

Description	Marks
Mitosis produces cells that replace the parent cells	1
so need to have the same structure and function to maintain the tissue or to heal a wound in the tissue	1
Total	**2**

Question 10

Description	Marks
Meiosis is not required for the survival of the individual,	1
but it is necessary for the survival of the species as it is the basis for the production of new individuals of the species	1
Total	**2**

CHAPTER 19: DNA

Question 1

Description	Marks
the concentration of the electrolyte solution	1
the density of the gel	1
the voltage used	1
the length of time the voltage is applied	1
Total	**4**

Question 2

Description	Marks
One peak – homozygous i.e. the two chromosomes have the same number of minisatellites	1
two peaks – heterozygous	1
Total	**2**

Question 3

Description	Marks
Short tandem repeat	1
Total	**1**

Question 4

Description	Marks
Restriction enzyme	1
Total	**1**

Question 5

Description	Marks
3'-TCGCTACATGCG-5'	1
Total	**1**

Question 6

Description	Marks
Hydrogen bonds allow the separation of the strands of DNA across the nucleotides with low energy input	1
will break well before the covalent bonds between the phosphates and deoxyribose sugars	1
Total	**2**

Question 7

Description	Marks
STR only requires small pieces of DNA	1
STR is fast and automated	1
STR uses small amounts of DNA that are amplified using the Polymerase Chain Reaction (PCR)	1
STR results are easily compared and stored	1
RFLP can take up to a month to accomplish	1
RFLP requires large amounts of non-degraded DNA	1
RFLP – automation is not possible	1
RPLF are stored as images, takes longer to compare and more storage space	1
Total	**8**

Question 8

Description	Marks
Minisatellites are small sequences of DNA	1
that do not encode proteins	1
appear throughout the genome hundreds of times, with many repeated copies lying next to each.	1
Individuals differ in the number, location and size of minisatellites on chromosomes making each person unique	1
Total	**4**

Question 9

Description	Marks
The 3' and 5' refer to the carbon atoms in the deoxyribose sugar. DNA polymerase	1
adds nucleotides to the chain at the 3' end due to the orientation of the phosphate group binding sites.	1
Total	**2**

Question 10

Description	Marks
It determines the movement of the fragments through the gel under the influence of an electric current	1
The DNA fragments are negatively charged and move towards the positive terminal in electrophoresis	1
The electrical charge causes the fragments to move in a predetermined direction which would not happen if movement was left to diffusion only	1
Total	**3**

CHAPTER 20: DNA REPLICATION AND MUTATION

Question 1

Description	Marks
Nucleotides are in the fluid contents of the nucleus.	1
Total	**1**

Question 2

Description	Marks
Ozarki fragments are formed from the replication of the lagging strand of DNA	1
where the nucleotides are connected in reverse order and in sections then the sections – called Ozarki fragments are joined together by DNA ligase to make a continuous strand.	1
Total	**2**

Question 3

Description	Marks
Substitution – the incorrect nucleotide presented at the connecting site	2
Deletion – the nucleotide was not quick enough to take its place in the sequence	2
Insertion – two nucleotides present at the same time.	2
Total	**6**

Question 4

Description	Marks
The genetic code relies on the correct sequence of nucleotides to produce the correct sequence of amino acids for protein structure and function.	1
Without the correct proteins the cellular functioning will be compromised e.g. enzymes require a specific structure to binds with the reactants to facilitate a reaction	1
Total	**2**

Question 5

Description	Marks
Some amino acids are coded for by several different triplet codons.	1
If the changed triplet still codes for the same amino acid, then no phenotypic change will occur.	1
Total	**2**

Question 6

Description	Marks
DNA ligase is essential for the joining of the Ozarki fragments during DNA replication.	1
If there is no DNA ligase, this doesn't happen and the DNA will not be replicated correctly (the DNA will be in pieces) causing the cells to die.	1
Total	**2**

CHAPTER 21: EPIGENETICS

Question 1

Description	Marks
(a) – Each twin is exposed to different types and amounts of epigenetic factors during their lifetime. – They are exposed to different environments with different amounts of methylation. – They have different probabilities of methylation occurring as this is a random event.	3
(b) – Tobacco smoke has not affected the cells/ DNA in a way to cause cancer in long-term smokers. – Methylation is a random event, even though it is increased by smoking, there are still chances that smoking will have no effect on cells to cause cancer	2
(c) – Older people have had greater exposure to sources of methylation in the environment over their lifetime than younger people. – The cumulative effects of methylation events could impact the gene expression more than individual events.	2
(d) – Methylation of embryonic stem cells could have a far greater impact than methylation of cells with lower potency levels, such as the differentiated cells of an adult.	2
Total	**9**

Question 2

Description	Marks
Changes in epigenome occur according to their individual exposure to different factors affecting phenotype.	1
Total	**1**

CHAPTER 22: PROTEIN SYNTHESIS

Question 1

Description	Marks
The sequence determines the attachments to other amino acids.	1
forming sheets or coils and attachment points for other protein chains in the tertiary and quaternary structure	1
Total	**2**

Question 2

Description	Marks
The DNA code uses thymine as the pair for adenine; mRNA uses uracil	1
Total	**1**

Question 3

Description	Marks
To make synthetic RNA of all the different combinations – one at a time, to see what amino acid chains form to link each amino acid to a specific codon	1
Total	**1**

Question 4

Description	Marks
STOP codons are recognized by the 'release factors' in the ribosomes,	1
and these trigger a reaction called hydrolysis, which causes the amino acid chain to be released from the ribosome and move out into the cytoplasm.	1
There is no tRNA with the complementary code to bring in another amino acid to join the chain	1
Total	**3**

Question 5

Description	Marks
met – pro – gly – his – val – leu – gin – leu – his – ser – leu – lys – ser – STOP	3
Total	**3**

Question 6

Description	Marks
T A C G G A C C A G T A C A T G A T G T T G A A G T A A G A A A T T T C A G A A T T	3
Total	**3**

Question 7

Description	Marks
T A C G G A C C A G T A C A T G A T G T T G A A G T A A G A A A T T T C A G A A T T	2
T A C G G A C C A G T A C A T G A T G T T G A A G T A A G A A A T T C A G A A T T	2
met – pro – gly – his – val – leu – gin – leu – his – ser – leu – lys – ser – leucine NO STOP codon	2
Total	**6**

Question 8

Description	Marks
The change in the shape of the alpha chain causes	1
an overall change in the shape of the haemoglobin	1
and the red blood cells making it less efficient at carrying oxygen.	1
Total	**3**

Question 9

Description	Marks
A protein's structure determines how it interacts with other chemicals.	1
e.g. enzymes need to fit into the active sites of the substrate for the reaction to occur.	1
Total	**2**

Question 10

Description	Marks
STRs are short tandem repeats of genetic code.	1
They do not code for anything therefore are classed as introns.	1
Total	**2**

CHAPTER 23: GAMETE FORMATION

Question 1

Description	Marks
(a) The acromere contains enzymes that breakdown the layer of cells surrounding the ovum to allow access for a single sperm to penetrate the membrane of the ovum.	1
(b) The midpiece contains large numbers of mitochondria that release the energy required to move the tail.	1
(c) The tail whips about to move the sperm forward	1
Total	**3**

Question 2

Description	Marks
The ova can be up to about 50 – 55 years old as women of this age group can still become pregnant because they are still releasing ova during their menstrual cycle if they have not gone through menopause.	2
Sperm are only about 3 months old at any time during the males life time as they degenerate after that time	2
Total	**4**

Question 3

Description	Marks
Sperm cells are much smaller than an ovum.	1
Length of sperm is about half the diameter of an ovum. The size of the head of the sperm is about 1/60th the diameter of the ovum.	2
Volume of an ovum is about 900,000 cubic microns: Volume of a sperm is about 65 cubic microns.	2
Total	**5**

Question 4

Description	Marks
The polar bodies degenerate and the materials reabsorbed into the body	1
Total	**1**

Question 5

Description	Marks
The zona pellucida regulates fertilization – only allows one sperm to enter the ovum and	1
keeps the ovum and cells from the first cell divisions together before implantation	1
Total	**2**

Question 6

Description	Marks
Corona radiata is a thick outer layer that surrounds the zona pellucida..	1
It is a cellular layer that is composed of proteins, carbohydrates and hyaluronic acid. It is formed by the adhesion of follicle cells	1
It protects and supplies vital proteins to the ovum	1
Total	**3**

CHAPTER 24: PEDIGREE CHARTS

Question 1

Description	Marks
(a) 4 and 5 heterozygous (Hh). Probability of each passing on the affected allele is 0.5 Probability of offspring receiving an affected allele from both parents is 0.5 x 0.5 = 0.25	3
(b) 9 and 10 each have 0.5 probability of being heterozygous (Hh). Each has 0.5 probability of passing on the H allele if they are heterozygous 0.5 x 0.5 = 0.25 Probability of 16 receiving H from each parent is 0.25 x 0.25 = 0.125	3
(c) Probability of producing a son is 0.5 Probability of producing a normal offspring is 0.75 Probability of producing a normal son is 0.75 x 0.5 = 0.375	3
Total	**9**

Question 2

Description	Marks
Autosomal recessive	1
autosomal – two alleles are required not one, therefore it is not sex-linked	1
recessive two alleles of the same type are required	1
Total	**3**

Question 3

Description	Marks
Both parents	1
Albinism requires two recessive alleles to have that phenotype.	1
One allele comes from each parent	1
Total	**3**

Question 4

Description	Marks
Autosomal – has two alleles controlling the inheritance of the characteristic	2
Recessive – only one allele (almond eyes) is required to have the phenotype of almond eyes	2
Total	**4**

Question 5

Description	Marks
2, 5 and 8 – each have colour blind sons.	1
3 – unknown as she has no children	1
7 – unknown as her only child is an unaffected female	1
Total	**3**

Question 6

Description	Marks
A female could get the disease only by having a mother who is a carrier	1
and a father who has the disease	1
Total	**2**

Question 7

Description	Marks
Father could be $I^a I^a$ or $I^a i$	1
Mother can only be ii	1
Offspring could be $I^a i$ or ii.	1
Total	**3**

Question 8

Description	Marks
Autosomal dominant	1
only requires one allele to have the affected phenotype	1
Total	**2**

Question 9

Description	Marks
Both parents are	1
Ff Each parent has probability of 0.5 for donating f to their child. 0.5 x 0.5 = 0.25	1
Each child from this combination of parents has 0.25 probability of having cystic fibrous	1
Total	**3**

Question 10

Description	Marks
(a) Probability of ?1 parents having the allele is 0.5 each. Probability of producing gametes carrying affected allele is 0.25 for each parent. Probability of ?1 having the condition is 02.5 x 0.25 x 0.125	3
(b) We do not know if you are a carrier of the condition or not; however, your child is a carrier	2
Total	**5**

Question 11

Description	Marks
Probability of III2 being a carrier is 0.5 x Probability III2 donating affected gamete to P is 0.5	2
Probability of P being a male is 0.5 = 0.125	2
Total	**3**

CHAPTER 25: MALE REPRODUCTIVE SYSTEM

Question	Answer	Mark
1	d	1
2	c	1
3	b	1
4	b	1
5	d	1
6	a	1
7	c	1
8	a	1
9	a	1

Question 10

Description	Marks
fructose – energy source for sperm	2
alkaline fluids – neutralise the acids of the vagina, mobility of sperm	2
Total	**4**

CHAPTER 26: FEMALE REPRODUCTIVE SYSTEM

Question 1

Description	Marks
Ovaries produce the hormones that make it possible for the female to mature and be able to produce offspring	1
Total	**1**

Question 2

Description	Marks
pathway for ovum to move from the ovary to the uterus	1
location of fertilisation	1
Total	**2**

Question 3

Description	Marks
GnRH activates the anterior pituitary to produce FSH.	1
FSH promotes the production of oestrogen from the ovarian follicle.	1
Increased concentration of oestrogen is detected by the hypothalamus	1
GnRH production by hypothalamus is reduced	1
This reduces the amount of FSH produced by the anterior pituitary	1
Total	**5**

Question 4

Description	Marks
Only one sperm is required to fertilise the ovum to produce the diploid number of chromosomes in the embryo.	1
More than one sperm would cause chromosomal problems that would cause the embryo to die	1
Total	**2**

Question 5

Description	Marks
Ovary – corpus luteum degenerates to become the corpus albicans, reducing the production of oestrogen and progesterone.	2
Uterus – low levels of oestrogen and progesterone cause the breakdown of the endometrium which ends with the menstrual flow	2
Total	**4**

Question 6

Description	Marks
in the current caused by fluids from ovulation and secretions from fallopian tubes	1
movement of the cilia lining the fallopian tubes	1
Total	**2**

Question 7

Description	Marks
The breakdown products of the endometrium including blood and epithelial tissue	1
Total	**1**

Question 8

Description	Marks
A. fallopian tube	1
B. ovary	1
C. uterus	1
D. cervix	1
E. vagina	1
Total	**5**

Question 9

Description	Marks
A. corpus luteum	1
B. carpus albicans	1
C. primary follicle	1
D. secondary follicle	1
E. ovum	1
Total	**5**

CHAPTER 27: CONTRACEPTION

No questions for this chapter.

CHAPTER 28: SEXUALLY TRANSMITTED INFECTIONS

Question	Answer	Mark
1	b	1
2	d	1
3	a	1
4	d	1
5	c	1
6	c	1
7	d	1
8	a	1
9	c,d,e,f	4

Question 10

Question	Answer	Mark
a	T	1
b	F	1
c	T	1
d	F	1
e	F	1
f	T	1
g	T	1
h	F	1

CHAPTER 29: CONCEPTION AND IMPLANTATION

Question 1

Description	Marks
A fertilised ovum formed at conception does not implant into the endometrium	1
and is flushed through the uterus and out of the body	1
Total	**2**

Question 2

Description	Marks
sperm acromere contains enzymes to breakdown the corona radiata and penetrate the zona pellucida	1
zona pellucida allows only one sperm to enter the ovum	1
Total	**2**

Question 3

Description	Marks
the enzymes from many sperm are required to breakdown the corona radiata	1
many sperm are damaged or poorly formed during spermatogenesis	1
many sperm die on the journey to the fallopian tube – run out of energy, or are overcome by the chemical environment of the female reproductive tract	1
many sperm may go into the other fallopian tube that does not contain the ovum	1
Total	**4**

Question 4

Description	Marks
There is not enough time for the fertilised ovum to go through the early embryonic stages before implanting into endometrium	1
Fluids currents will take it out of the uterus before it can be implanted	1
Total	**2**

Question 5

Description	Marks
Two or more embryos can develop as the cells at these stages	1
as all cells are totipotent and differentiation has not started	1
Total	**2**

Question 6

Description	Marks
The blastocysts has an uneven distribution of cells in the structure..	1
The location of the cells and cell surroundings could determine which cells differentiate into different things	1
Total	**2**

CHAPTER 30: ASSISTED REPRODUCTIVE TECHNOLOGIES (ART)

Question 1

Description	Marks
Low sperm count (Too few or no sperm in the semen)	1
Low sperm motility (Sperm don't move as well as they should)	1
Malformation of the sperm	1
Blocked sperm ducts	1
Total	**4**

Question 2

Description	Marks
True	1
Men and women are equally likely to have a fertility problem. In about one in five infertile couples, both partners have contributing problems	1
Total	**2**

Question 3

Description	Marks
False	1
IVF is a complex and expensive procedure; only about 5% of couples with infertility seek this treatment.	1
Total	**2**

Question 4

Description	Marks
True	1
Obesity can inhibit regular ovulation and may increase the risk of complications during pregnancy such as gestational diabetes, hypertension, preeclampsia and respiratory problems	1
Total	**2**

Question 5

Description	Marks
False	1
There may be many structural or functional or hormonal problems that cause infertility that are not related to general overall health.	1
Total	**2**

Question 6

Description	Marks
False	1
It depends on the state of the eggs, the age of the woman and the problems to overcome by using IVF	1
Total	**2**

Question 7

Description	Marks
True	1
A study found that a woman's fertility starts declining gradually starting in her late 20s. Although the decline in the 20s is probably clinically insignificant, it does become significant by the early 30s. Most women in their late 20s or early 30s can become pregnant fairly easily, but it might take a month or two longer than it would have in their early 20s. Postponing pregnancy until a woman is in her late 30s or her 40s is likely to be trickier, though. According to the Mayo Clinic, age is the biggest predictor of fertility – the older a woman is, the harder it will be to conceive. Fertility takes a sharp decline by age 37 and over the age of 45, the odds of getting pregnancy using your own eggs is less than 1 percent (although women undergoing fertility treatments may be able to get pregnant using donor eggs).	1
Total	**2**

Question 8

Description	Marks
False	1
According to the American Society of Reproductive Medicine, 85% to 90% of all cases of infertility can be treated with fertility drugs or other 'conventional' medical therapies	1
Total	**2**

Question 9

Description	Marks
True	1
common guidelines used by most fertility clinics	1
Total	**2**

Question 10

Description	Marks
False	1
On average, menopause begins 1.5 years earlier in women who smoke. It has also been suggested that the effect of smoking on female fertility is similar to a 10-year increase in age	1
Total	**2**

CHAPTER 31: GENETIC TESTING AND EMBRYONIC SCREENING

Question 1

Description	Marks
female – 2 X chromosomes	1
Down's syndrome – 3 chromosome 21s – producing cognitive impairment,	1
phenotype – flattened face, small head, short neck, protruding tongue, upward slanting eye lids, unusually shaped or small ears	1
Total	**3**

Question 2

Description	Marks
Amniocentesis takes samples of the amniotic fluid surrounding the foetus.	1
CVS takes samples of the villi of the placenta	1
Total	**2**

Question 3

Description	Marks
Can cause rupture of the placenta and bleeding which could lead to nutrient supply problems with the foetus and, at worst, miscarriage	1
Injury or infection to the foetus	1
Limb deformity if done too early	1
Leaking amniotic fluid into the surrounding tissue	1
Total	**4**

Question 4

Description	Marks
Testing for genetic abnormalities – chromosomal or single gene disorders e.g. cystic fibrosis infection	1
maternal-foetal blood compatibilities	1
growth defects such as heart, neural tube and cleft palate	1
normal development and growth	1
Total	**4**

Question 5

Description	Marks
nothing – continue with the pregnancy	1
terminate the pregnancy	1
Total	**2**

Question 6

Description	Marks
non-disjunction of chromatids during meiosis	1
where both chromosome 21s move to the same end of the dividing cell	1
Total	**2**

CHAPTER 32: CELL POTENCY AND CANCER

Question 1

Description	Marks
Self-renewal: Stem cells are capable of continuous cell division	1
Potency: Stem cells are capable of differentiation (are effectively unspecialised precursors to the production of specific types of cells)	1
Total	**2**

Question 2

Description	Marks
Pluripotent – can form any cell type (but can't form extra-embryonic structures like the membranes forming the placenta)	1
Multipotent – can differentiate into closely related cell types	1
Total	**2**

Question 3

Description	Marks
Embryonic stem cells – from the inner cell mass of the blastocyst stage of the embryo	1
Adult stem cells – bone marrow, breasts, intestines, fat tissue, brain, nose, hair follicles and testes	1
Total	**2**

Question 4

Description	Marks
Mitosis producing extra cells causing excess number of cells (tumour)	1
not controlled by factors in their environment	1
Total	**2**

Question 5

Description	Marks
Embryonic stem cells come from the blastocyst	1
which is destroyed during the process – destruction of a viable human embryo that could have grown into a healthy child	1
Total	**2**

Question 6

Description	Marks
Leukemia is when bone marrow produces an excessive amount of abnormal white blood cells, which don't function properly.	1
Radiation kills these cells	1
Bone marrow transplant inserts bone marrow stems cells that produce the normal blood cell types	1
Total	**3**

Question 7

Description	Marks
Increasing age gives the cells more time to turn faulty or mutate and grow into cancers	1
Prolonged exposure to carcinogens such as sunlight, radiation, environmental chemicals (smoking), and substances in the food eaten including alcohol increases the risk of developing cancers.	1
Cells accumulate more mutations the longer we live, increasing the risk of developing cancers	1
Total	**3**

Question 8

Description	Marks
cells from the original tumour break away and from the cell mass that is usually surrounded by a membrane	1
enter the blood or lymph	1
carried around the body	1
moved out of the blood or lymph into other tissue	1
uncontrolled mitosis of these cells forms new tumours	1
Total	**5**

CHAPTER 33: EMBRYONIC DEVELOPMENT AND BIRTH

Question	Answer	Mark
1	a	1
2	d	1
3	b	1
4	d	1
5	b	1
6	c	1
7	a	1
8	b	1
9	d	1
10	c	1

NOTES

Titles available from Academic Group

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STUDY GUIDES - YEAR 11 & 12

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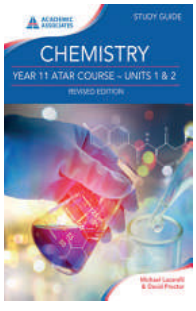

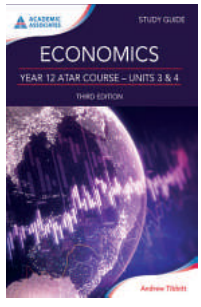

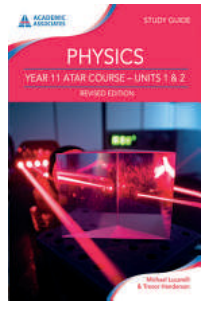

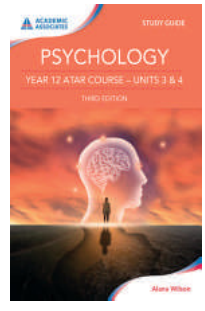

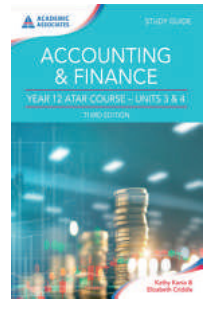

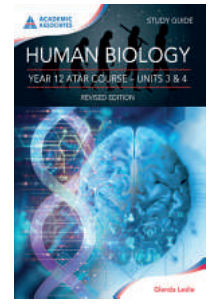

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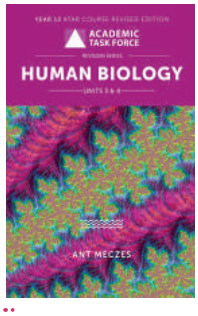
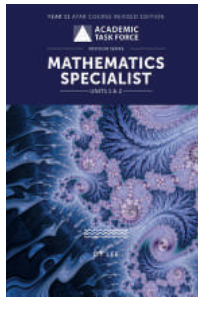
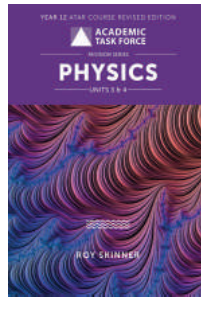
Physics | Psychology

TEXT BOOKS - YEAR 11 & 12

Mathematics Methods | Mathematics Specialist







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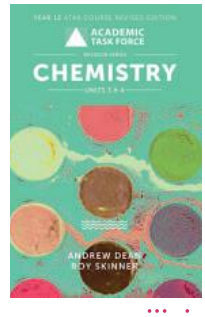

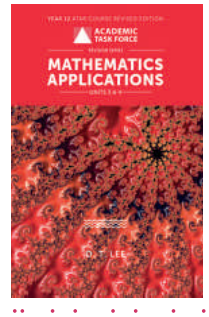

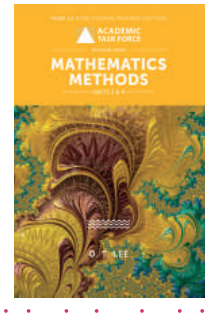

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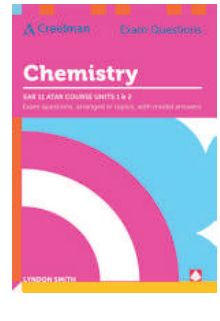

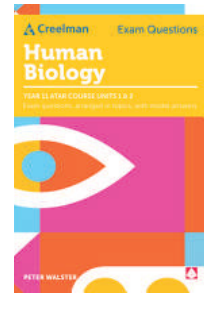

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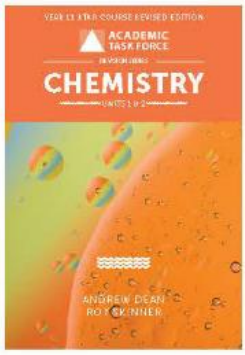

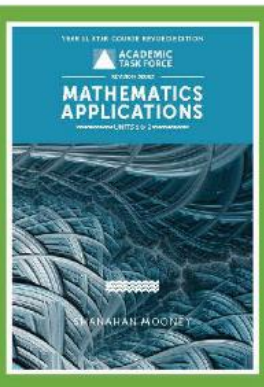

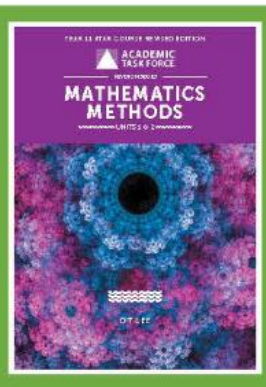

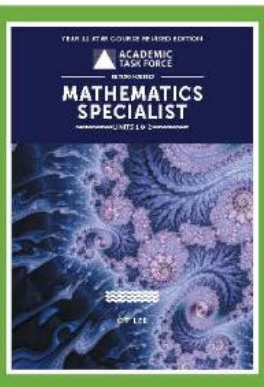

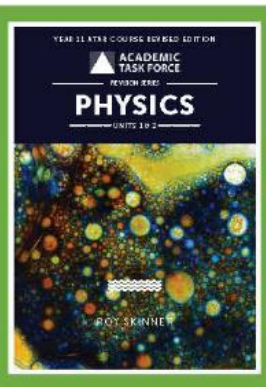

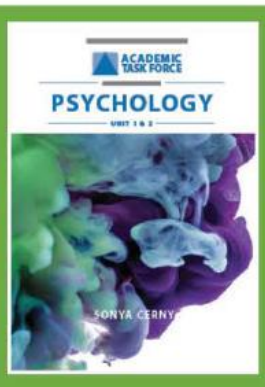

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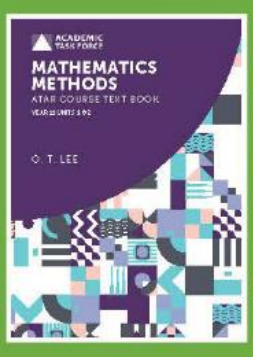

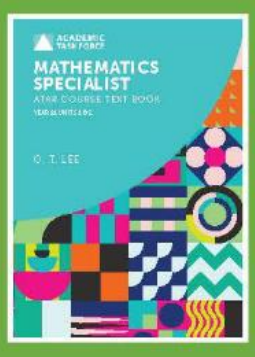

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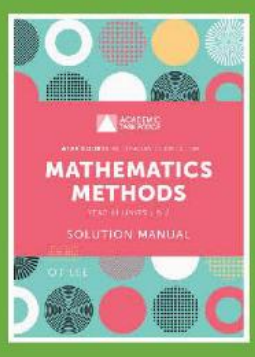

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