WORKBOOK

SECOND EDITION
Rhys Lewis

SACE STAGE 2
Australian Curriculum

Biology

SASTA
South Australian Science Teachers Association

1.2: Genes and protein synthesis

A gene consists of a unique sequence of nucleotides that codes for a functional protein or an RNA molecule.

Protein synthesis involves transcription of a gene into messenger RNA (mRNA), and translation of mRNA into an amino acid sequence at the ribosomes. In eukaryotic cells, transcription occurs in the nucleus.

- Describe and illustrate the role of DNA, mRNA, transfer RNA (tRNA), ribosomal RNA (rRNA) in transcription and translation.
- Describe the relationship between codons, anticodons, and amino acids.
- Distinguish between coding (gene) and template strands of DNA.
- Distinguish between exons and introns as coding and non-coding segments of DNA found in genes.
- Describe how both exons and introns are transcribed, but only the information contained in exons is translated to form a polypeptide in eukaryotes.

Genes are sequences of hundreds to millions of DNA nucleotides that code for the assembly of a functional **protein** or RNA molecule. Genes in eukaryotes contain sequences that code for a protein, called **exons**, as well as sequences that do not code for a protein, called **introns**.

Proteins are large molecules that are essential to cell structure and function. A protein is composed of smaller molecules called **amino acids**, and the sequence of amino acids in a protein is coded for by the sequence of DNA nucleotides in a gene. Proteins are synthesised from genes in several stages that are summarised in Figure 1.10.

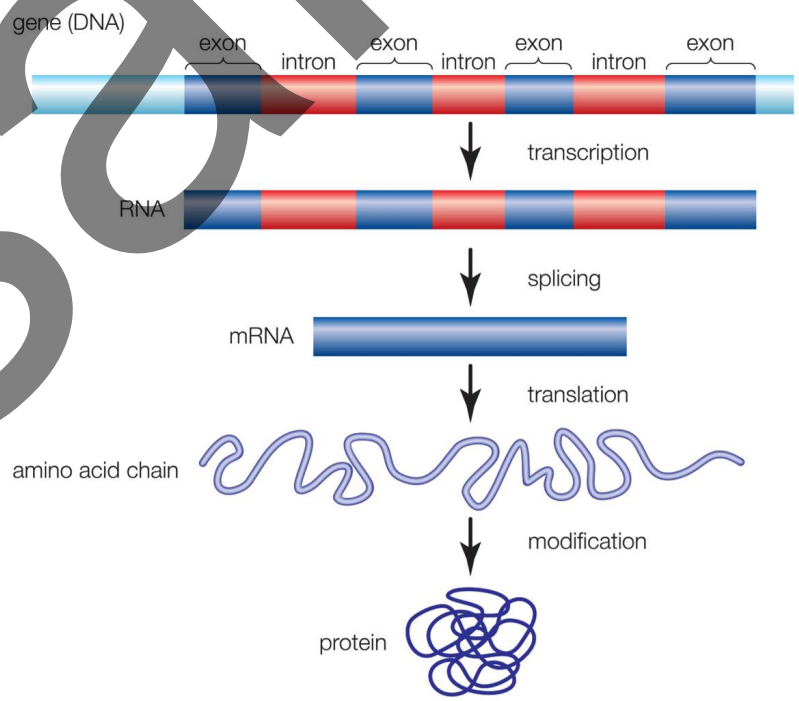


Figure 1.10: Stages of protein synthesis.

STR analysis

A Short Tandem Repeat (STR) analysis is a method used in biology to compare the short tandem repeat sequences at specific loci on the DNA from two or more samples. Scientists have identified several loci in the human genome where STRs are commonly found, and some are identified below.

Chromosome	Locus	STR sequence
2	TPOX	AATG
3	D3S1358	TCTA TCTG TCTA
5	CSF1PO	ATCT
7	D7S820	TATC
10	D10S1248	GGAA
11	THO1	TCAT
16	D16S539	GATA

The number of STR repeats at certain loci is highly variable, and this property allows forensic scientists to compare the DNA of two individuals.

The data from STR analysis is used to construct an **STR electropherogram** which is a graphical representation of a DNA profile showing the locations of STRs on certain chromosomes. Figure 1.68 is an STR electropherogram showing the locations of STRs on four different human chromosomes.

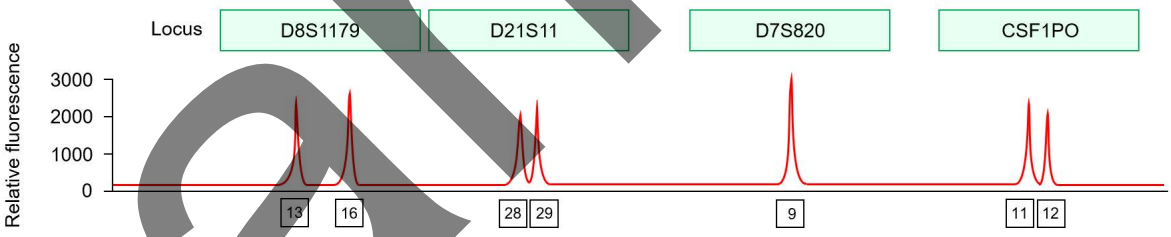


Figure 1.68: STR electropherogram.

The STR electropherogram shows two peaks at D8S1179, D21S11, and CSF1PO, indicating that the individual is heterozygous at these loci. The presence of two separate peaks at these loci is due to the alleles on the paternal and maternal chromosomes of a homologous pair having different numbers of short tandem repeat sequences. For example, the individual has an allele containing an STR with 13 repeats and one with 16 repeats at locus D8S1179. In contrast, the presence of one peak at D7S820 indicates that the individual is homozygous at this locus as the STR is the same length on the paternal and maternal chromosomes.

STR electropherograms are used to compare the genotypes of two individuals in DNA profiling, and the results may be used as forensic evidence or to determine paternity, maternity and heredity.

Example 1.11

The probability of two individuals having the same number of STRs at a given locus is very low, and the probability decreases as more loci are compared. This property allows forensic scientists to rule out suspects based on similarities with DNA samples recovered from a crime scene. Figure 1.69 is an electropherogram comparing DNA from a crime scene with that of two suspects. The DNA profile from the crime scene matches Suspect B as all of their alleles are present in the sample.

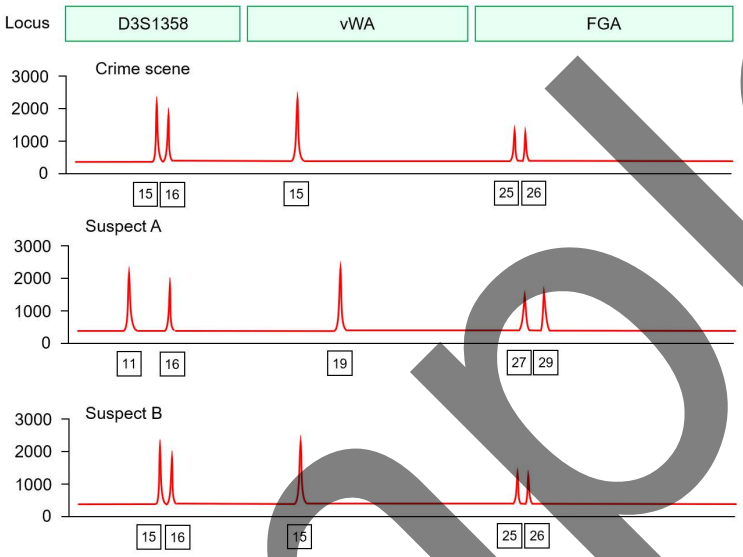


Figure 1.69: STR electropherogram from a crime scene.

Example 1.12

STR electropherograms are also used in paternity and maternity testing. Children inherit STRs from their parents which allows for the identification of paternity and maternity by comparing DNA profiles. Figure 1.70 is an electropherogram that identifies Elizabeth as the daughter of Henry and Anne as one allele from each pair of STRs in Elizabeth comes from Henry and the other from Anne.

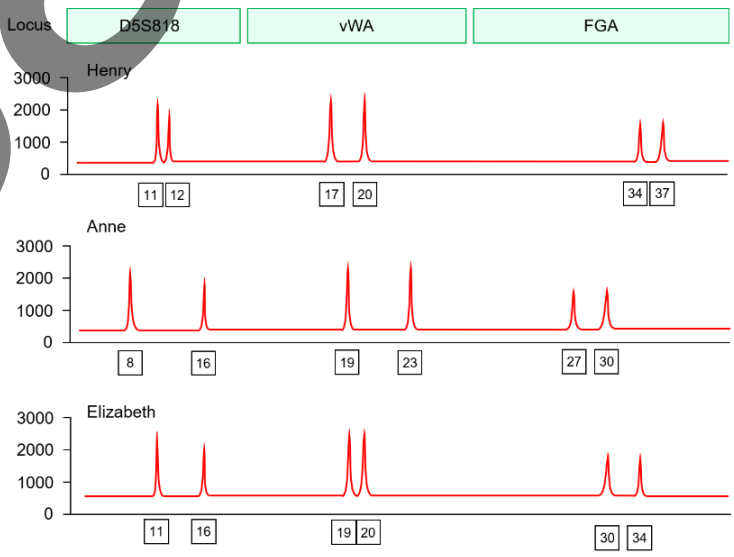


Figure 1.70: STR electropherogram showing paternity and maternity.

DNA profile table

The data from an STR electropherogram can be displayed in a **DNA profile table** (also called an Autosomal STR DNA profile). The numbers in the table are called **allele values** which identify the number of repeats in the STRs on the alleles of an individual at certain loci. The information in a DNA profile table is used in the same way as an STR electropherogram and can be used to determine paternity, maternity, family lineage and to identify a suspect using DNA found at a crime scene.

Example 1.13

The DNA profile table below shows the allele values of a mother, her child and the alleged father.

Locus	Alleged father	Child	Mother
AMEL	X, Y	X, X	X, X
D8S1179	15.3, 14	15.3, 17	15, 17
D3S1358	16, 17.2	12, 16	14.3, 12
D13S317	11,13	13,14	14, 16
D7S820	10,12	8, 12	8, 10
TPOX	9, 14	7, 14	7, 9
CSF1PO	12, 15	10, 12	8, 10
D21S11	30,31	28, 30	28, 29
vWA	18, 18	16, 18	15, 16

The table shows that the alleged father is the likely parent of the child as one allele from each pair of STRs in the child is present in the alleged father.

Example 1.14

The DNA profile table below shows the allele values of two suspects and DNA found at a crime scene.

Locus	Crime scene	Victim	Suspect A	Suspect B
D3S1358	15, 16, 18	15, 15	12, 14.3	16, 18
THO1	6,8,9.3	6, 9.3	10, 12	6, 8
D21S11	28, 29, 32.2	28, 29	26, 28.2	29, 32.2
D18S51	13, 14, 16	13, 14	11.1, 13	13, 16
D5S818	11, 12, 13	11, 13	9, 10.1	11, 12
D13S317	10, 11, 12	11, 12	13, 14	10, 11
vWA	17, 18	18, 18	18, 20	17, 17

The table shows that there is evidence that Suspect B was at the crime scene as both alleles from each pair of STRs in Suspect B are present in the DNA obtained from the crime scene.

Question 37

The DNA profile table below contains data obtained from a crime scene, a victim and two suspects.

Locus	Crime scene	Victim	Suspect A	Suspect B
D3S1358	15, 17, 19	15, 19	15, 17	16, 17
THO1	9, 11, 12	9, 11	9, 12	7,9
D21S11	29, 30, 31, 32.2	31, 32.2	29, 30	28, 32
D18S51	12, 16, 18	12, 16	12, 18	10, 12
D5S818	11, 12	11, 12	11, 11	10, 13
D13S317	11, 13	11, 11	11, 13	9, 11
vWA	15, 17, 18	17, 18	15, 17	13, 16.1
FGA	21, 23	21, 23	23, 23	20, 21

THO1 is a locus on human chromosome 11 that contains the short tandem repeat sequence AATG.

(a) Define a short tandem repeat.

_____

_____ (1 mark) **KA1**

(b) The victim has the DNA profile below.

Locus	Allele values
THO1	9, 11

(1) State the evidence that the victim is heterozygous at THO1.

_____ (1 mark) **KA1**

(2) Explain what the numbers 9 and 11 represent in the table above.

_____

_____

_____

_____ (2 marks) **KA1**

(c) State and explain which suspect was present at the crime scene.

_____

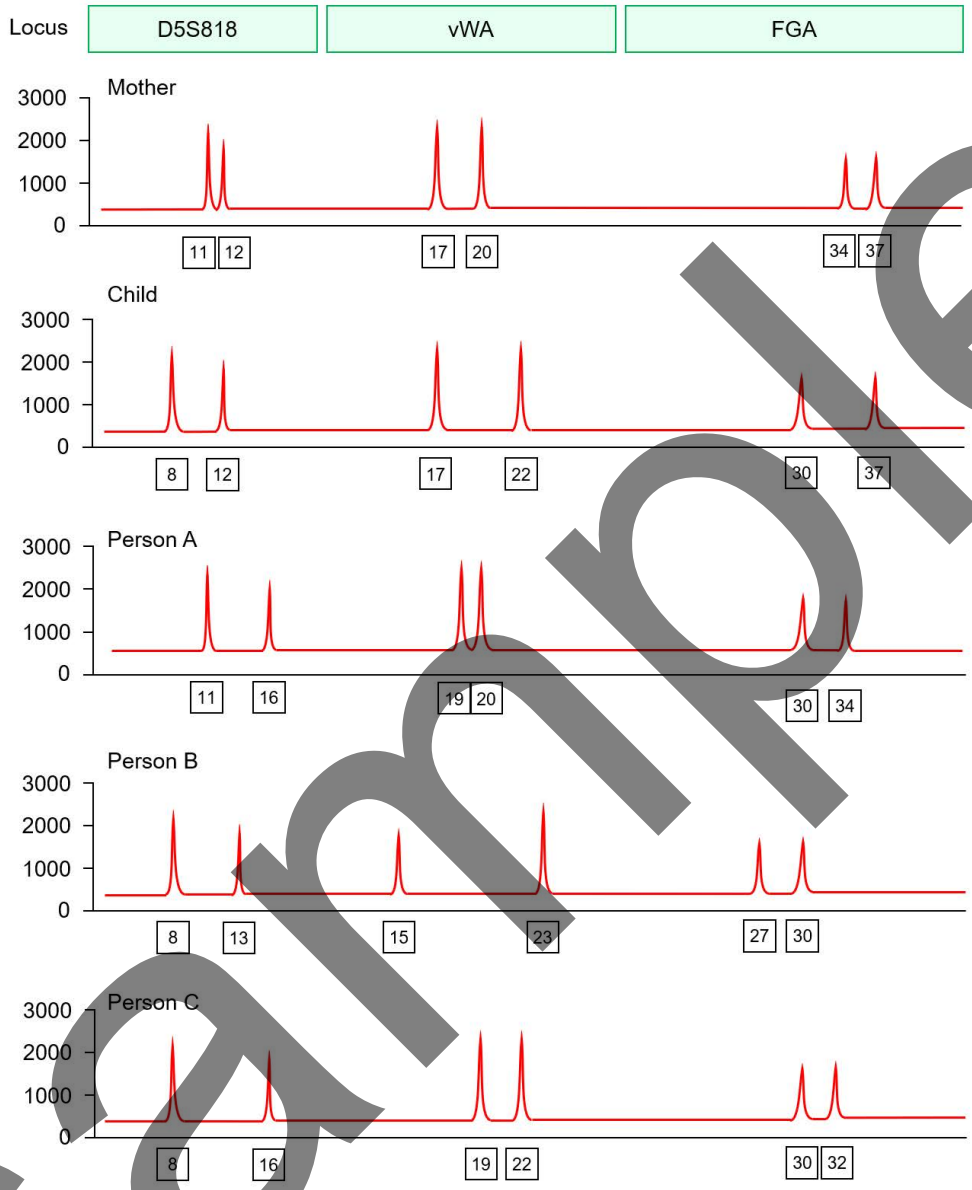
_____

_____

_____ (2 marks) **KA2**

Question 39

The diagram below is an STR electropherogram used to identify the paternity of a child.



(a) Describe and explain how the information in the STR electropherogram is used to determine the paternity of the child.

(2 marks) **KA2**

(b) Identify the father of the child using the information in the electropherogram.

(1 mark) **KA4**

Protein design

Protein design is the science of developing new protein molecules to control biochemical processes in cells for applications including biomedical research, medicine and technology. Proteins can be designed from scratch or redesigned by making specific modifications to existing proteins. Modern protein design involves the use of computer software to predict the three-dimensional structure and folding of a protein from its amino acid sequence. The designed proteins may then be synthesised using biotechnology techniques. Some applications and examples of designed proteins are described in the table below.

Application	Description
Enzyme design	Scientists design enzymes that bind with a substrate with high specificity. These enzymes are used in commercial applications, including the food industry and biomedical research.
Studying protein-protein interactions	Scientists design proteins that interact with proteins involved in the development and progression of disease. It is hoped that the designed protein could bind to and inhibit the disease-causing protein.
Development of new materials	Scientists design proteins with desirable physical and chemical properties such as high tensile strength, improved toughness and durability, and high ductility.
Targeted chemotherapy	Scientists design proteins that prevent tumour growth by interfering with specific target molecules needed for the replication of cancer cells. Other proteins are designed to attach to the membrane of cancer cells where they attract immune cells that destroy the tumour.

Biosensors

Biosensors are designed receptor proteins, antibodies, or enzymes that detect chemical substances in their environment by binding to target molecules with high specificity. There are many potential applications of biosensors, and some examples are identified below.

Application	Example
Medical	Monitoring blood glucose levels in diabetics.
Environmental	Detecting pesticides and river water contaminants
Counter-bioterrorism	Sensing airborne bacteria during a biological attack.
Disease prevention	Detection of pathogens in a host or environment.
Food quality	Detection of contaminants in food products.
Food safety	Detection of toxic metabolites in foods such as mycotoxins.

Review Test 1

Question 1

Place a cross in the box next to the answer you think is most correct.

(a) A DNA molecule contains 180,000 guanine bases and 220,000 thymine bases.

What is the total number of nucleotide bases in the DNA molecule?

J. 180,000
K. 220,000
L. 800,000
M. 400,000

(1 mark)

(b) The table below shows some mRNA codons and their amino acids.

mRNA codon	Amino acid
ACC	Threonine
AUU	Isoleucine
CUU	Leucine
UCC	Serine

A tRNA molecule has the anticodon AGG

Which amino acid is attached to the tRNA molecule?

J. Threonine
K. Serine
L. Isoleucine
M. Leucine

(1 mark)

(c) Rett syndrome is a neurological disorder caused by mutations in the MeCP2 protein which regulates gene expression.

Which one of the following is **not** a mechanism of silencing genes?

J. Increased methylation of cytosine nucleotides in DNA
K. Increased activity of microRNAs and siRNAs
L. Increased activity of transcription factors and repressor proteins
M. Increased conversion of heterochromatin to euchromatin

(1 mark)

Question 4

Catalase is an enzyme in liver cells that converts hydrogen peroxide to oxygen.

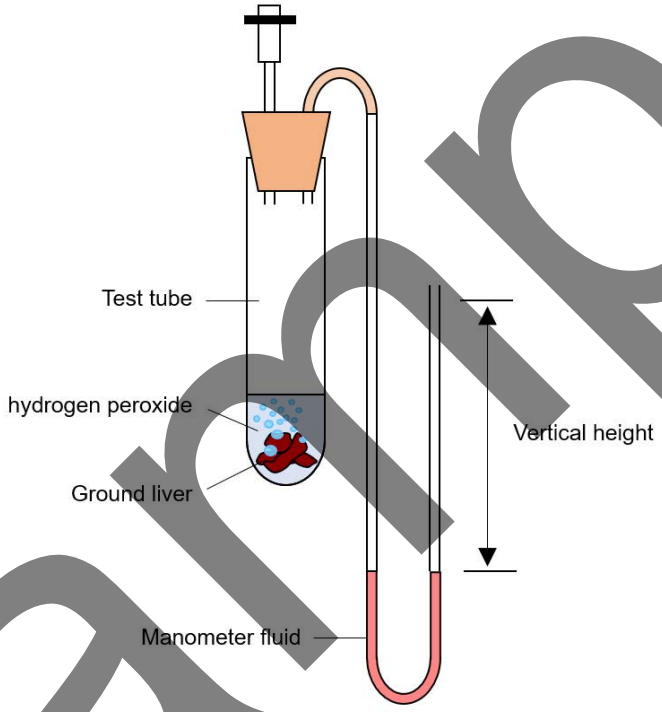
An investigation was conducted to determine the effect of temperature on the activity of catalase.

Ground samples of liver tissue were transferred to five separate beakers containing pure water at different temperatures until they reached thermal equilibrium.

The samples were transferred to test tubes containing 5 mL of hydrogen peroxide.

The reaction produced oxygen which caused the fluid to travel vertically within the manometer.

The vertical distance travelled by the fluid was measured and recorded after 30 seconds.



(a) State one hypothesis for this investigation.

_____

_____ (1 mark) **IAE1**

(b) State the independent variable in this investigation and give one reason for your answer.

_____

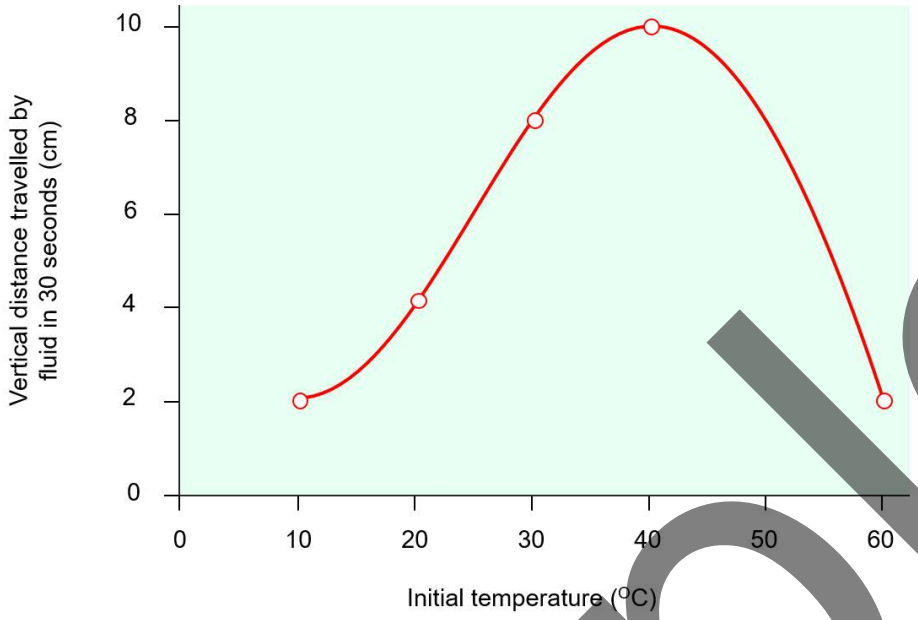
_____ (2 marks) **IAE1**

(c) State why it was necessary to maintain a constant pH in this investigation.

_____

_____ (1 mark) **IAE1**

(d) The graph below shows the pattern of results of the investigation.



(1) Describe and explain the pattern shown by the results of this investigation.

(3 marks) **IAE3**

(2) Identify one source of random error in this investigation.

(1 mark) **IAE4**

(3) State why it is important to minimise the effect of random error in the investigation.

(1 mark) **IAE4**

Question 5

Refer to the following information:

> CRISPR gene editing is a technique used in biotechnology to edit the genomes of living things. The technique is based on the CRISPR-Cas9 antiviral defence system in bacteria that was first described by researchers at Osaka University in 1987.
>
> Since that time, more than 1 000 research papers about CRISPR have been published by scientists at universities and research facilities around the world, and the gene-editing technique has already been used in clinical trials with varied results. It is hoped that the results of clinical trials will inform scientists and allow them to improve treatments and minimise harmful side-effects for patients undergoing treatment with CRISPR.
>
> CRISPR technology has been used to edit genes in human cells, to modify microorganisms to make biofuels, to modify plants and improve crop yields, and has the potential to treat a wide range of genetic diseases, including cancer, sickle cell disease, haemophilia, cystic fibrosis, Huntington's, and heart disease.
>
> Scientists are currently investigating the use of CRISPR-Cas3 to modify mosquitos so they cannot transmit diseases such as malaria and to destroy viruses inside the human body including HIV, herpes, and HPV. Research is also being conducted into the use of Cas-12a to modify plants.

Discuss how this example demonstrates one or more key aspects of science as a human endeavour.

(6 marks) **KA3**

The cell membrane

The **cell membrane**, also called the **plasma membrane**, is a thin boundary that separates a cell from the extracellular environment. The function of the cell membrane is to control the movement of materials, including raw materials and wastes, between the cell and its environment. The cell membrane has a thickness of approximately eight nanometres and consists of a bilayer of **lipids,** including **sterols** and **phospholipids** as shown in Figure 2.02.

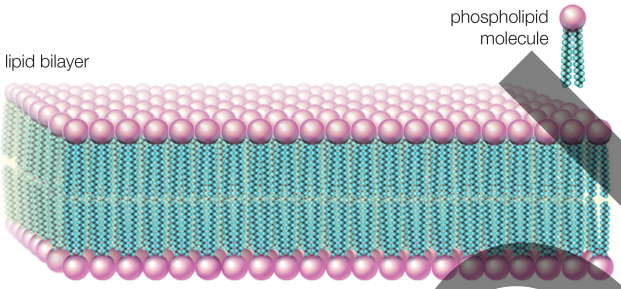


Figure 2.02: Phospholipid bilayer of the cell membrane.

The cell membrane also contains a variable amount of protein. Some proteins are embedded in the membrane, and others span the width of the membrane. The cell membrane is described using the **fluid-mosaic model,** which states that the membrane is fluid, as individual phospholipids move within and between layers, and contains a mosaic of embedded proteins (Figure 2.03).

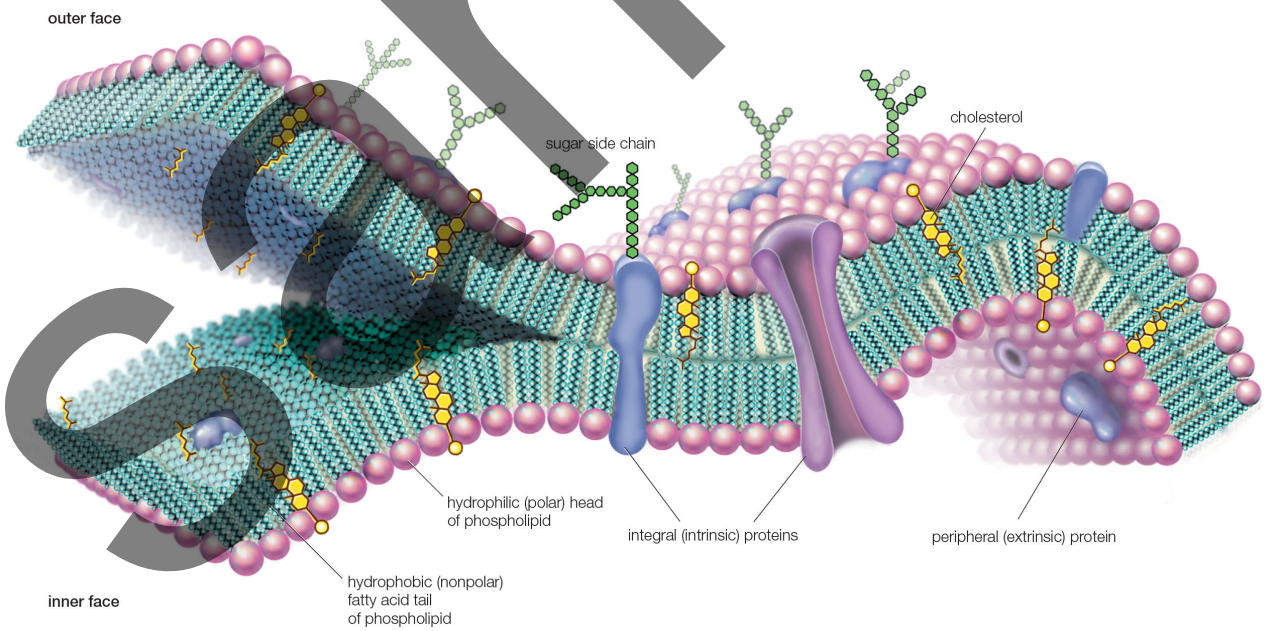


Figure 2.03: Fluid mosaic model of the cell membrane.

The cell membrane is composed of lipids and proteins. Lipids are a family of chemical compounds with similar physical properties which includes fats, phospholipids, and cholesterol. The three types of lipids in the membrane of eukaryotes are phospholipids (75%), sterols (20%), and glycolipids (5%).

Endoplasmic reticulum (ER)

The **endoplasmic reticulum** (Figure 2.17) is a continuous membrane system that serves multiple functions, including the synthesis, folding, modification, and transport of proteins. There are two distinct types of endoplasmic reticulum that have different structures and functions. The **rough ER** has **ribosomes** attached to its outer surface which gives it a rough appearance. The membrane of the rough ER is continuous with the nuclear envelope which allows mature mRNA molecules to diffuse out of the nucleus following transcription and RNA splicing and attach to ribosomes for translation. In contrast, the **smooth ER** has no ribosomes and is involved in the synthesis of lipids, including cholesterol and phospholipids used in the membranes of the cell and its organelles.

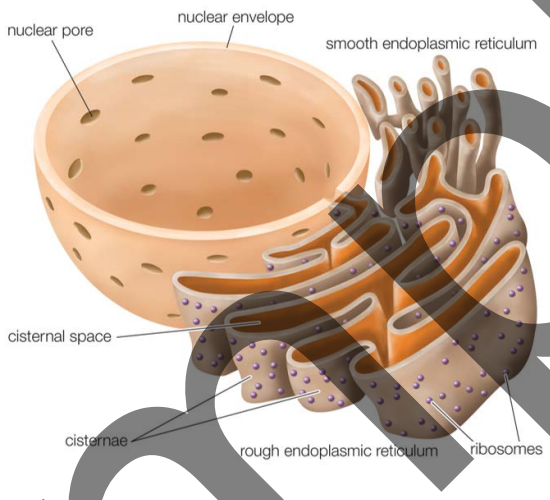


Figure 2.17: Structure of the endoplasmic reticulum (ER).

Golgi Body

The **Golgi body,** also called the **Golgi apparatus** (Figure 2.18), is a membrane-bound organelle located next to the endoplasmic reticulum and near the cell nucleus that is made up of a series of flattened pouches called cisternae. The Golgi body is responsible for transporting, modifying, and packaging proteins and lipids into **vesicles** for delivery to targeted destinations.

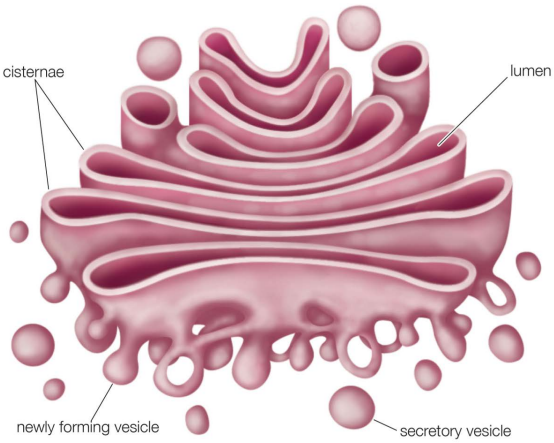
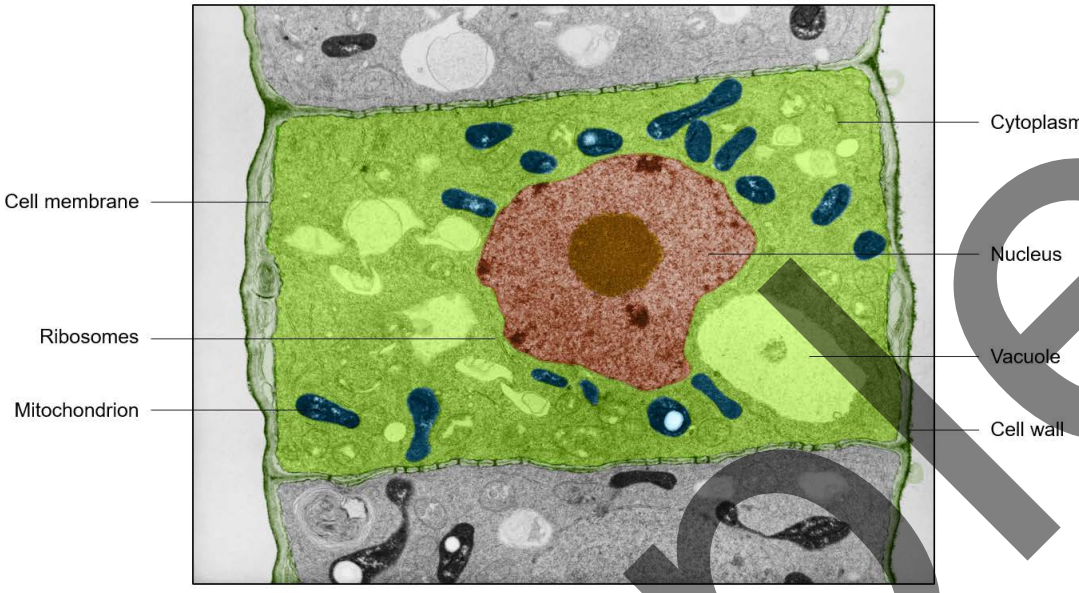


Figure 2.18: Structure of a Golgi body.

Question 49

The diagram below is a coloured TEM of an onion root cell.



(a) Identify two features of this cell that are also present in a prokaryotic cell.

_____ (2 marks) **KA1**

(b) Identify the two features of this cell that are not present in a prokaryotic cell.

_____ (2 marks) **KA1**

(c) State one feature of this cell that is not present in an animal cell.

_____ (1 mark) **KA1**

(d) Cells in the leaves of onions contain chloroplasts.

(1) State the primary function of chloroplasts.

_____ (1 mark) **KA1**

(2) Draw a labelled diagram of a chloroplast.

(2 marks) **KA1**

(3) State the reason why chloroplasts are not present in the cell in the diagram above.

_____

_____ (1 mark) **KA2**

The sun is the main source of energy for life.

- Recognise that photosynthesis is important in the conversion of light energy into chemical energy, as illustrated by the following equation:

$$6CO_2 + 6H_2O \xrightarrow[\text{chlorophyll}]{\text{light}} C_6H_{12}O_6 + 6O_2$$

$$\text{carbon dioxide} + \text{water} \xrightarrow[\text{chlorophyll}]{\text{light}} \text{glucose} + \text{oxygen}$$

The sun is the principal source of energy for living things on Earth. Energy is transferred from the sun to the Earth in the form of electromagnetic radiation (light), and this energy is absorbed by autotrophs such as plants, phytoplankton, and cyanobacteria that use the energy to carry out **photosynthesis**. Photosynthesis is a complex chemical process that converts light into chemical energy through the transformation of carbon dioxide and water into oxygen and sugars such as glucose as depicted in Figure 2.30.

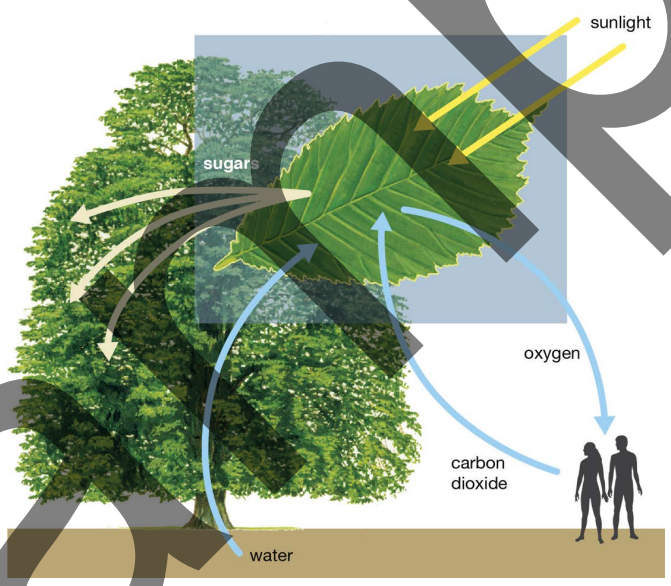


Figure 2.30: Photosynthesis

Autotrophs contain light-absorbing molecules such as **chlorophyll** that are stored in chloroplasts (eukaryotes) or thylakoid membranes (prokaryotes) located near the cell membrane. The light energy absorbed by autotrophs in photosynthesis is stored in chemical bonds between the atoms in molecules of glucose. The transformation of carbon dioxide and water into glucose and oxygen in photosynthesis is summarised using the symbol and word equations below.

$$6CO_2 + 6H_2O \xrightarrow[\text{chlorophyll}]{\text{light}} C_6H_{12}O_6 + 6O_2$$

$$\text{carbon dioxide} + \text{water} \xrightarrow[\text{chlorophyll}]{\text{light}} \text{glucose} + \text{oxygen}$$

Interphase

Interphase is the longest phase and accounts for approximately 90% of the duration of the cell cycle. Interphase is divided into subphases called G_1, S, and G_2, and the events occurring in each subphase are described in the table below.

Phase	Description
G_1	In G_1 phase (first gap phase), the cell grows in size and carries out metabolic reactions including protein synthesis and respiration. In addition, the cell accumulates DNA nucleotides and ATP in preparation for DNA replication in S phase.
S	In S phase (synthesis phase), the chromosomes are replicated, and the two copies of a chromosome are joined by a centromere. In addition, the cytoskeleton is dismantled, and the centrioles are replicated in preparation for mitosis.
G_2	In G_2 phase (second gap phase), protein synthesis continues, some organelles are replicated, ATP is synthesised, and the cell grows in size in preparation for M phase. In addition, the DNA is checked, and any damage is repaired before the cell enters M phase.

The cell cycle control system

The sequential events of the cell cycle are directed by a **cell cycle control system** that is regulated by both internal and external factors. The cell cycle is driven by specific chemical signals present in the cytoplasm and evidence for this comes from experiments in which mammalian cells at different phases of the cell cycle were fused to form a single cell with two nuclei. The results showed that a cell in G_1 phase immediately entered S phase when fused with a cell in S phase, and a cell in G_1 phase immediately entered M phase when fused with a cell in M phase (Figure 2.78).

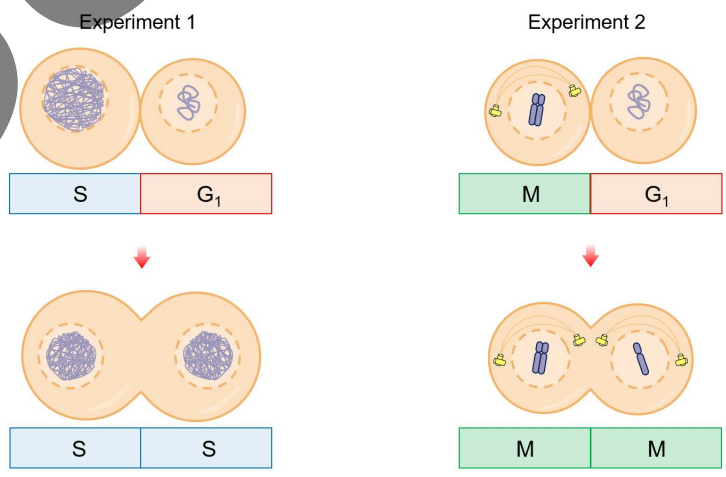


Figure 2.78: The cell cycle in eukaryotes.

Checkpoints

The cell cycle has specific **checkpoints** where the cycle is stopped, and the cell is prevented from entering the next stage if one or more conditions are unfavourable. The cell cycle is controlled by three internal checkpoints that are described in Figure 2.79 and the table below.

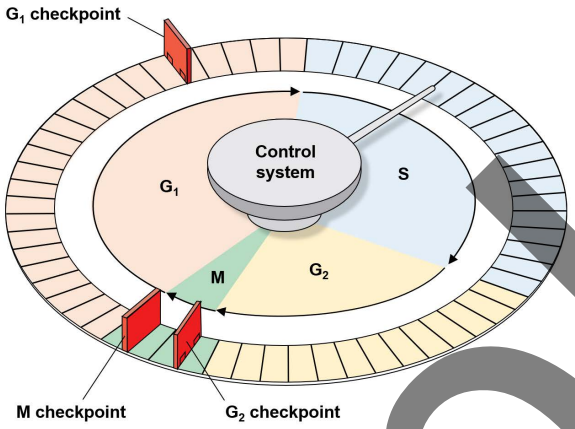


Figure 2.79: The cell cycle checkpoints in eukaryotes.

Checkpoint	Description
G_1	The availability of DNA nucleotides and ATP is assessed, and the chromosomes are checked for mutations and other forms of damage in preparation for S phase.
G_2	The genome is checked to ensure that all chromosomes have been replicated and that each chromosome is free from mutations and damage before the cell enters M phase.
M	The sister chromatids are checked to ensure that the centromere is attached to two spindle fibres before the chromatids are irreversibly separated in anaphase.

In many cases, the cell will typically complete the other phases of the cell cycle if it receives a go-ahead signal at the G_1 checkpoint. However, if the cell does not receive the go-ahead signal, it will exit the cycle and enter a non-dividing state called G_0 phase as depicted in Figure 2.80.

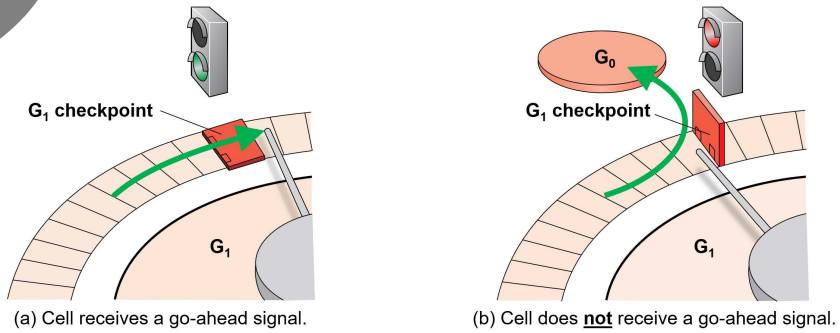


Figure 2.80: The cell cycle at the G_1 checkpoint.

3.1: Tolerance limits of organisms

Organisms survive most effectively within their tolerance limits.

Factors for which organisms have tolerance limits include:

- body temperature
- water availability
- blood glucose level
- carbon dioxide concentration in the blood and tissues.

There are impacts on an organism when conditions fall outside its tolerance limits.

There are millions of species of living things on Earth, including plants, animals, and microorganisms. The geographical distribution of each species is not uniform as each is adapted to live under different environmental conditions including temperature, water availability, nutrient availability, pH and salinity.

Tolerance limits

Organisms survive most effectively within **tolerance limits** for a given abiotic factor. Tolerance limits represent the maximum, and minimum amounts of a given factor a species can tolerate before the efficiency of life processes are reduced. The **tolerance range** represents the difference between the minimum and maximum tolerance limits to a given abiotic factor. Organisms with high tolerance ranges are widely distributed on Earth, whereas those with narrow tolerance ranges are confined to a smaller number of geographical locations and areas inside the body of a host.

Body temperature

Organisms regulate body temperature to ensure that metabolism occurs at a rate that maintains life processes. The metabolic reactions occurring in cells are catalysed by enzymes that each have an optimum temperature. The table below identifies consequences for enzyme activity when body temperature is outside the tolerance limits of an organism.

Temperature	Consequence
Below tolerance limit	The rates of enzyme-catalysed metabolic reactions are too slow to maintain the life processes in the cell.
Above tolerance limit	The shapes of the active sites of enzymes are altered, which decreases the rates of enzyme-catalysed metabolic reactions.

In humans, the tolerance range for body temperature is between 36 and 38°C. Controlling body temperature within narrow tolerance limits allows an organism to function effectively despite changes in the environmental temperature. The process by which organisms regulate body temperature is called **thermoregulation** and this concept is explored in Topic 3.2.

Water availability

Water is a chemical compound that is essential to all life processes, including metabolism, growth, movement, reproduction, and excretion. Organisms have mechanisms that regulate **osmolarity,** which refers to the relative concentrations of water and solutes in cells and tissues. The cells, tissues, and organs of living things operate in a fluid environment, and osmolarity must be maintained within fairly narrow tolerance limits. The table below identifies consequences for organisms when osmolarity is outside the tolerance limits.

Osmolarity	Consequence
Below tolerance limit	Cells increase in volume and become damaged as water diffuses into cells by osmosis. An increase in the volume of water may result in mild to serious health problems.
Above tolerance limit	Cells decrease in volume and become damaged as water diffuses out of cells by osmosis. The loss of water affects the folding of enzymes and proteins, which leads to serious health problems.

The normal blood osmolarity in humans ranges from 275 to 310 mOsm/kg, and this tolerance range is tightly regulated. The process by which organisms regulate solute concentrations and balance the gain and loss of water is called **osmoregulation,** and this concept is explored in Topic 3.2.

Blood glucose level

Glucose is the primary substrate for respiration in animals and most other living things. In animals, blood glucose levels are carefully controlled to ensure that a regulated quantity of glucose is available to body cells for respiration. The table below identifies consequences for the human body when the blood glucose level is outside tolerance limits.

Blood glucose level	Consequence
Below tolerance limit	A person becomes **hypoglycaemic** and has difficulty functioning as body cells are not receiving enough glucose to maintain respiration. If untreated, a person can lose consciousness.
Above tolerance limit	A person becomes **hyperglycaemic** and experiences complications, including excessive urination, thirst, and hunger. If untreated, complications from hyperglycaemia may be fatal.

The normal blood glucose level in humans is between 70 and 110 mg per 100 mL of blood which is known as the **fasting level** as it is measured after approximately 10 hours of fasting and represents the quantity of glucose being transported to the cells to maintain the rate of respiration. The regulation of blood glucose level is explored in Topic 3.2.

3.2: Stimulus-response and Negative feedback

Organisms detect and respond to changes in the internal and external environment.

Homeostasis is the maintenance of a relatively constant internal environment. This ensures the optimum conditions for the body to function.

In human beings, homeostasis depends on the functioning of the nervous and endocrine systems.

Homeostasis involves a stimulus–response and negative feedback model.

- Describe the role of sensory receptors.
- Describe the role of effectors.
- Explain the stimulus-response model.
- Recognise that in negative feedback, the response inhibits the initial stimulus.

Living things function most efficiently when the internal environment is reasonably constant. The process of maintaining a constant internal environment is known as **homeostasis**. Organisms use homeostasis to maintain constant internal conditions regardless of changes in the external environment. In humans, body temperature, osmolarity, glucose concentration, and blood pH are each maintained at a constant level. Homeostasis depends on the functioning of the nervous and endocrine systems and involves both **stimulus-response** and **negative feedback** models.

Stimulus-response model

The stimulus-response model is a mechanism of homeostasis. In the stimulus-response model, a change in an internal condition acts as a stimulus that is detected by **sensory receptors**. Sensory receptors respond to a stimulus by sending nerve impulses to a **control centre** such as the brain that interprets sensory input and coordinates the appropriate response. The control centre then sends nerve impulses or hormones to **effectors** such as muscles and glands that facilitate a response. The stimulus-response model of homeostasis is summarised in Figure 3.01.

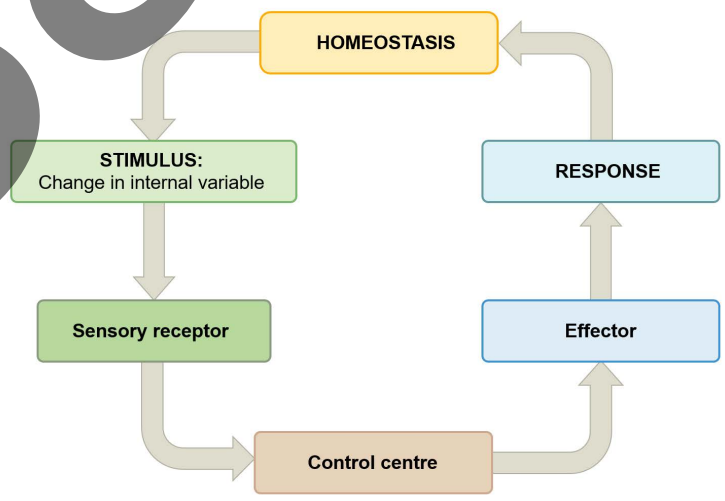


Figure 3.01: The stimulus-response model of homeostasis.

Sensory receptors

A homeostatic mechanism begins with sensory reception, which is the detection of a stimulus by sensory receptors. Sensory receptors are usually epithelial or nerve cells (neurons) located in sensory organs that detect stimuli both inside and outside the body. Sensory receptors translate the physical and chemical properties of stimuli into patterns of nerve impulses that are transmitted to the brain. Figure 3.04 shows two common types of sensory receptors in the human body.

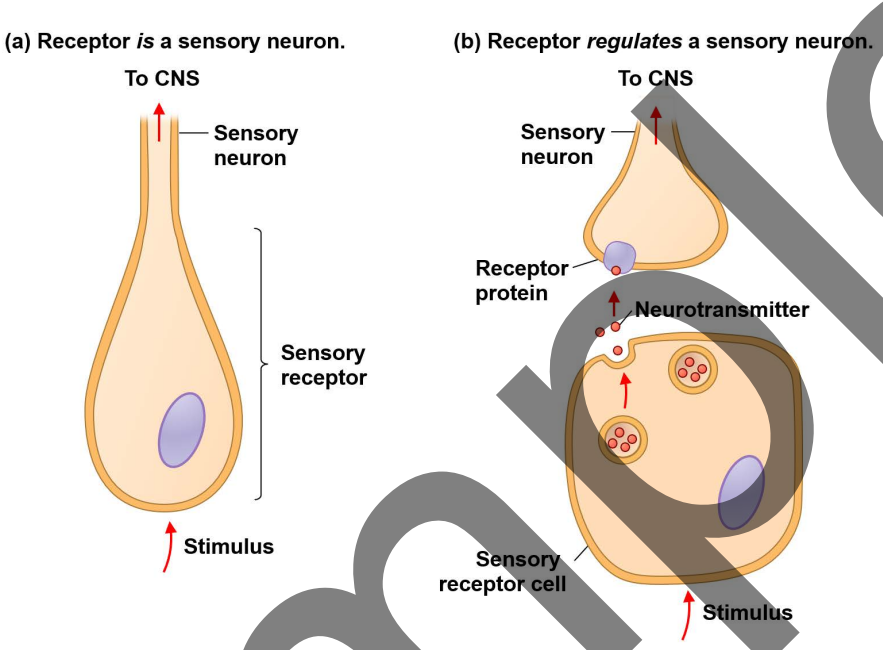


Figure 3.04: Sensory receptor cells in the human body.

When a stimulus is detected, sensory receptors convert the stimulus energy to a nerve impulse that is transmitted to the control centre for processing. Sensory receptors are classified based on the type of energy transduced, and the four common receptor types are described below.

Category	Description
Mechanoreceptors	Mechanoreceptors sense physical deformation caused by stimuli such as pressure, touch, stretch, motion, and sound.
Electromagnetic receptors	Electromagnetic receptors detect electromagnetic energy such as light (photoreceptors), electricity, and magnetism.
Thermoreceptors	Thermoreceptors sense changes in temperature in both the internal and external environment.
Chemoreceptors	Chemoreceptors respond to changes in the concentrations of chemicals caused by stimuli such as smell and taste as well as responding to changes in pH caused by carbon dioxide and osmolarity caused by the gain and loss of water.
Nociceptors	Nociceptors detect painful stimuli that could damage animal tissues and triggering defensive reactions.

Effectors

An effector is any tissue or organ that receives information from the control centre and facilitates the response required to maintain homeostasis. Some examples of effectors are identified below.

Regulatory process	Effector	Response
Thermoregulation	Skeletal muscles	Cause shivering to increase body temperature
Osmoregulation	Pituitary gland	Secretes ADH to increase absorption of water
Glucoregulation	Pancreas	Secretes insulin to decrease blood sugar level
Chemoregulation	Lungs	Increase or decrease the rate of ventilation

Negative feedback model

Homeostasis is maintained by **negative feedback**, a control mechanism that helps return a variable to the tolerance range by reducing the original stimulus. Most homeostatic control systems function by negative feedback, especially those that use hormones to regulate a given variable.

Example 3.01

Body temperature is affected by changes in the environmental temperature. Thermoreceptors in the skin, blood, brain and other sensory organs respond to a decrease in body temperature by sending nerve impulses to the hypothalamus in the brain. The hypothalamus stimulates the pituitary gland to secrete more thyroid-stimulating hormone (TSH) which stimulates the thyroid gland to secrete the hormone thyroxine (T_4). Thyroxine travels in the blood to tissue cells, where it increases the rate of heat-generating metabolic reactions. The response is an increase in body temperature which reduces the initial stimulus, as shown in Figure 3.05.

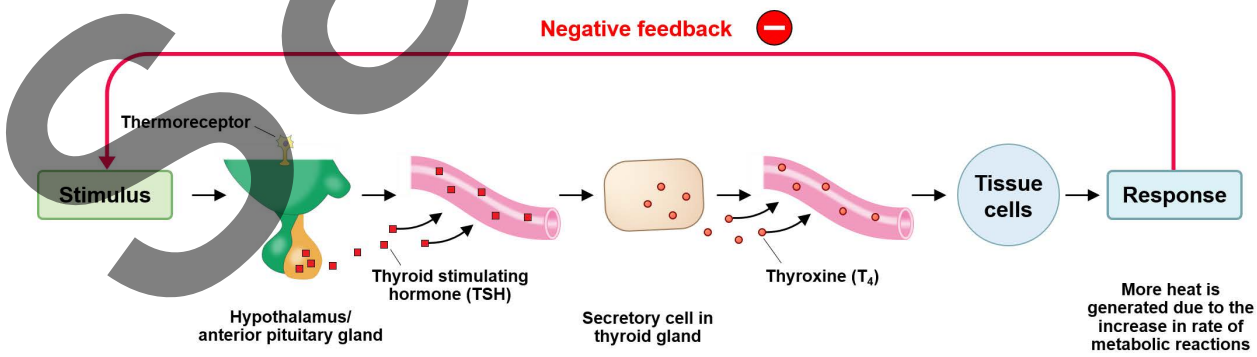


Figure 3.05: Negative feedback in the regulation of body temperature.

Thermoregulation

Thermoregulation is the process by which humans and other animals maintain an internal temperature within tolerance limits. Thermoreceptors in the body detect changes in the internal temperature and send nerve impulses to the hypothalamus in the brain, which acts as the control centre for thermoregulation. The hypothalamus stimulates effectors to carry out processes that either generate or remove heat from the body depending on the stimulus. Thermoregulation involves stimulus–response and negative feedback models that are summarised in Figure 3.08.

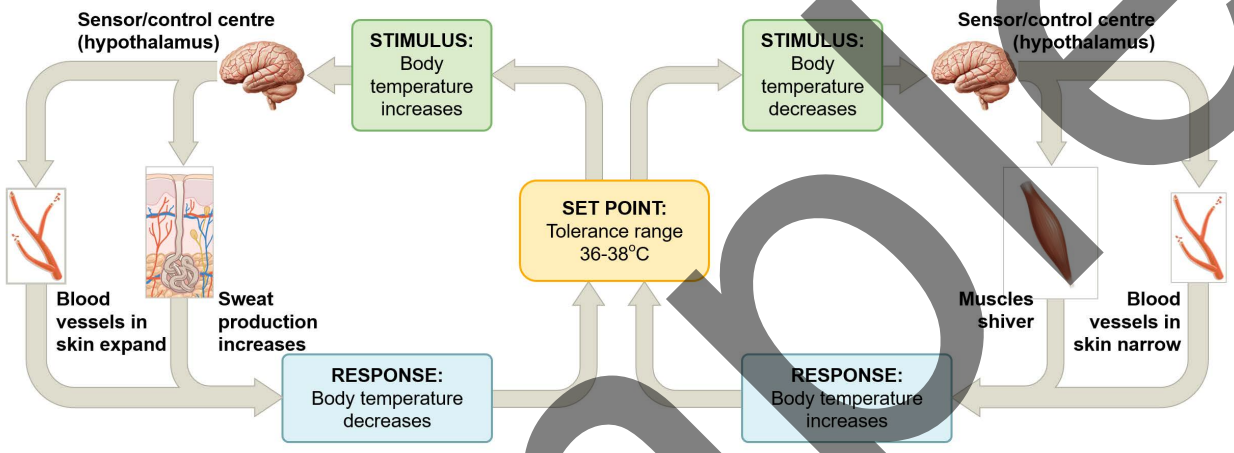


Figure 3.08: Thermoregulation in humans.

Osmoregulation

Osmoregulation is the process by which humans and other animals maintain osmolarity within tolerance limits. Osmoreceptors in the hypothalamus detect changes in osmolarity of the blood and stimulates the pituitary gland to increase or reduce the secretion of antidiuretic hormone (ADH) depending on the stimulus. Osmoregulation involves stimulus–response and negative feedback models that are summarised in Figure 3.09.

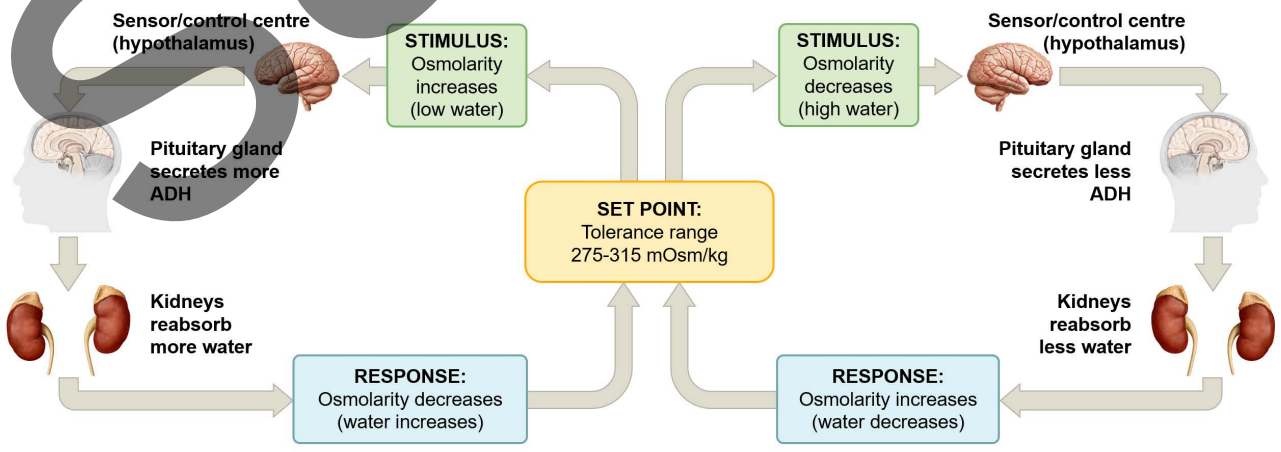


Figure 3.09: Osmoregulation in humans.

Glucoregulation

Glucoregulation is the regulation of blood sugar levels in humans and other animals. Chemoreceptors detect changes in blood sugar levels and stimulate the pancreas to secrete the hormones insulin and glucagon depending on the stimulus. Glucoregulation involves stimulus–response and negative feedback models that are summarised in Figure 3.10.

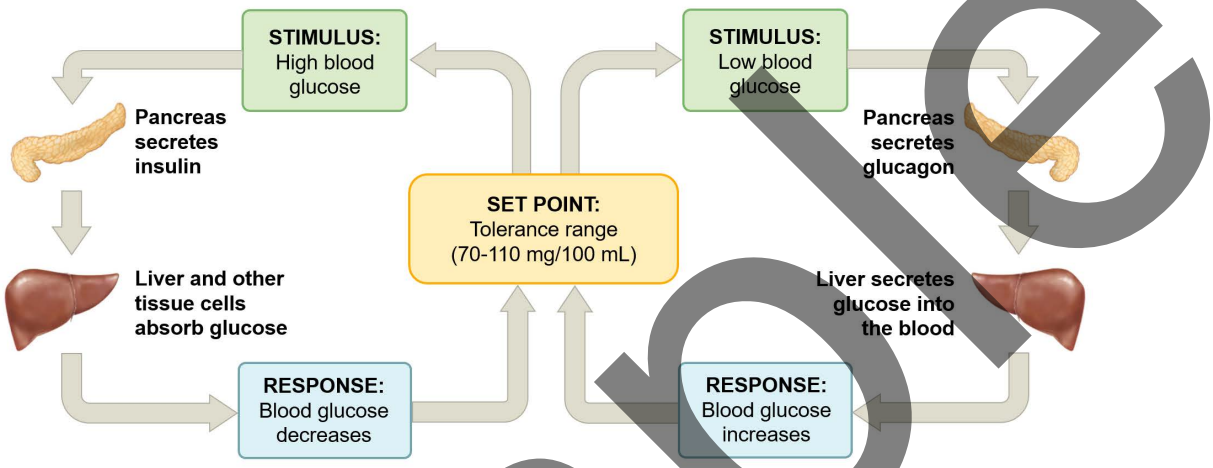


Figure 3.10: Glucoregulation in humans.

Chemoregulation

Chemoregulation is the regulation of blood pH which changes due to the presence or absence of respiratory carbon dioxide in the blood. Chemoreceptors in the brain and heart detect changes in blood pH and stimulate the medulla oblongata to change the rate and depth of ventilation depending on the stimulus. Chemoregulation involves stimulus–response and negative feedback models that are summarised in Figure 3.11.

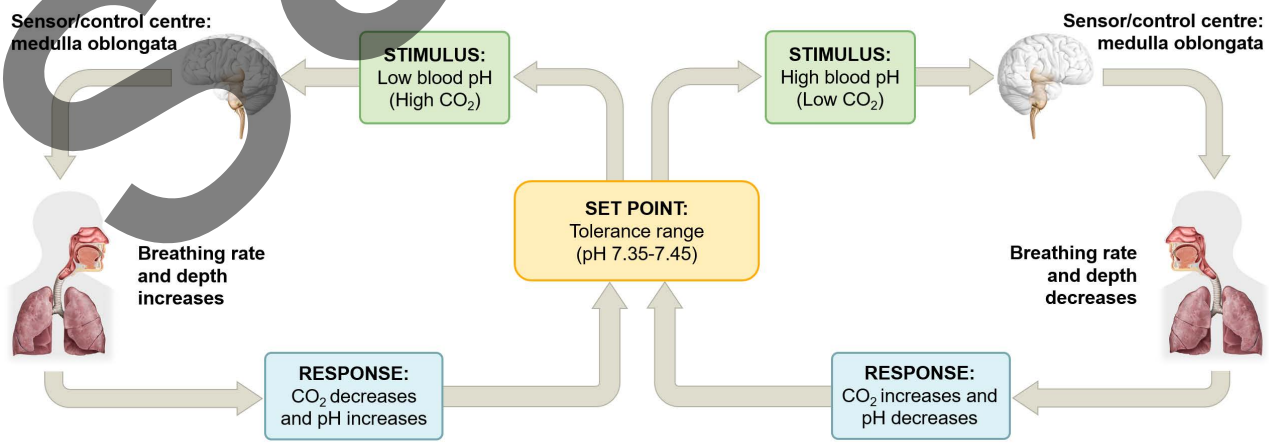


Figure 3.11: Chemoregulation in humans.

Neurons

Neurons are the primary cell type in the nervous system. Neurons have a highly specialised structure that enables the rapid transmission of nerve impulses between sensory receptors, the control centre, and effectors. Figure 3.15 shows the structure of a neuron.

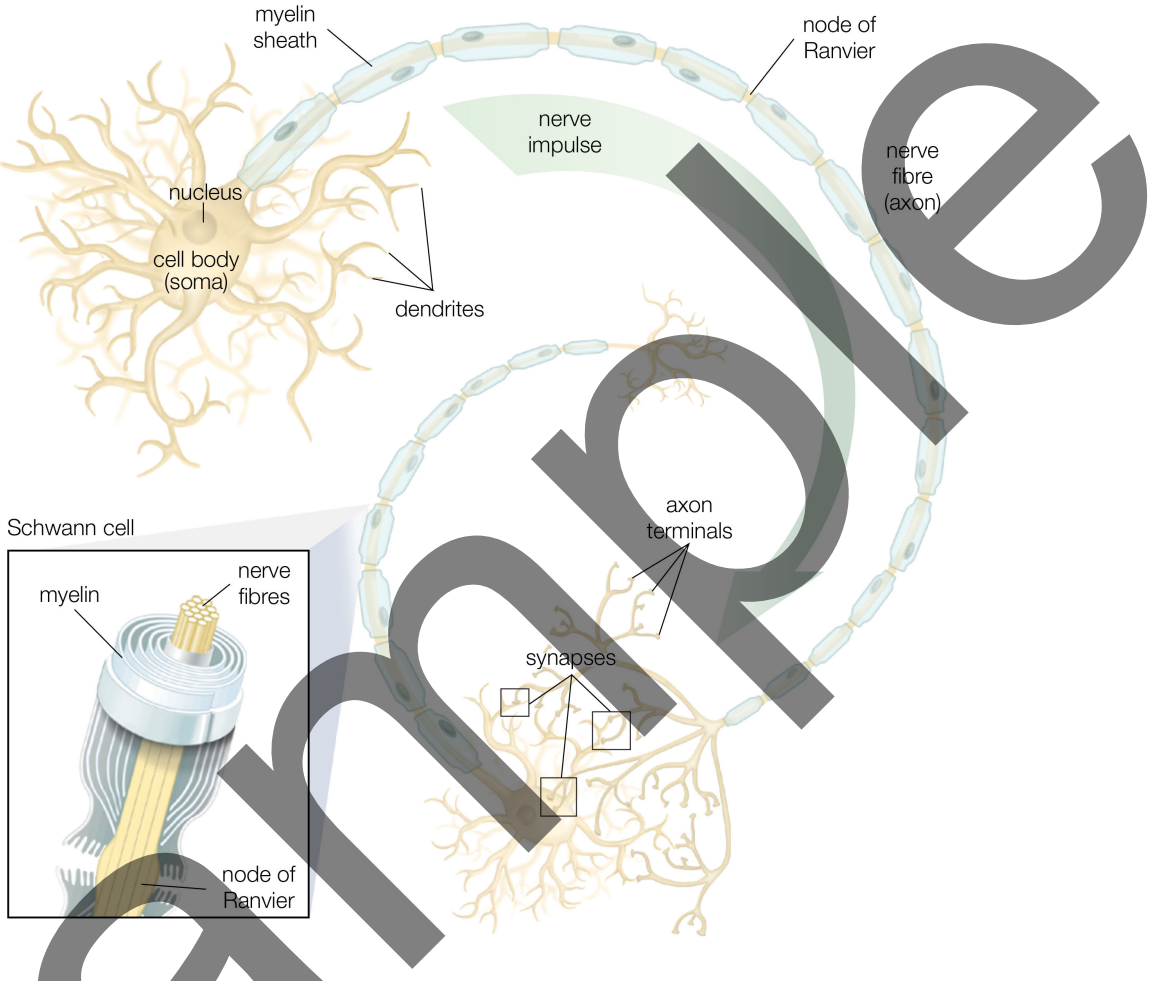


Figure 3.15: Structure of a neuron.

The table below shows the functions of the primary structural features of a neuron.

Feature	Function
Cell body (soma)	Contains the organelles that facilitate life processes in the cell.
Dendrites	Extensions of the cell body that receive nerve impulses from other cells.
Axon	A long fibre that conducts nerve impulses from the cell body to synapses.
Myelin sheath	A protective layer of support cells that insulate the axon.
Axon terminals	The branches of the axon that transmit nerve impulses to other neurons.
Synapse	The region between two nerve cells in a neural pathway.

Types of neurons

The peripheral nervous system contains sensory receptors that detect a stimulus and transmit the information to the central nervous system. The central nervous system interprets the information and transmits nerve impulses to effectors that facilitate the appropriate response to the original stimulus. The transmission of nerve impulses between the central and peripheral nervous systems occurs via three distinct types of neurons called sensory neurons, interneurons, and motor neurons. Figure 3.18 shows differences in the structure of the three functional types of neuron.

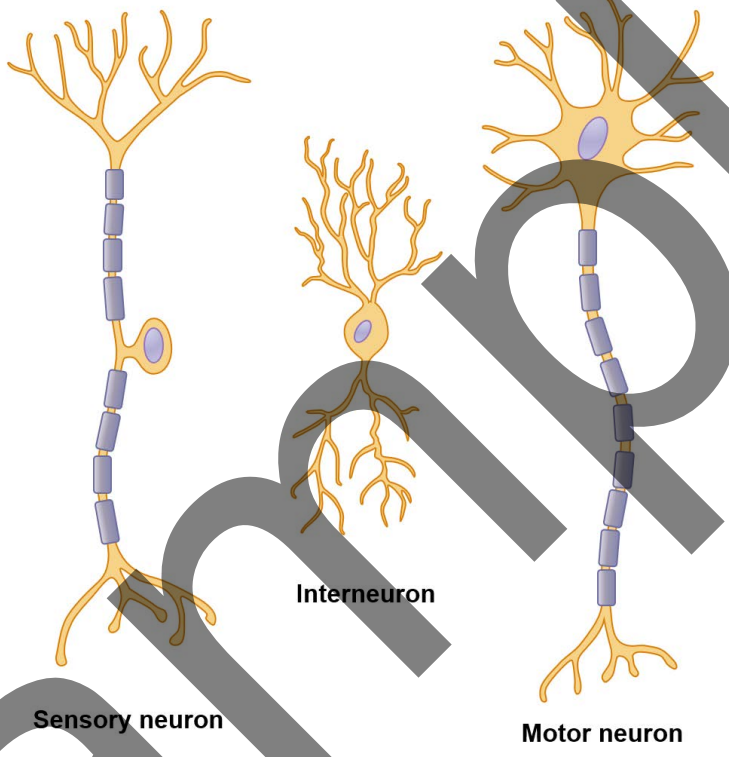


Figure 3.18: Three functional types of neuron.

The table below shows the location, structural features, and function of the three types of neuron.

Cell type	Location	Structural features	Function
Sensory neuron	PNS	• Long axon • Myelinated or unmyelinated • Cell body on the side of axon	Conduct nerve impulses from sensory receptors to the CNS.
Interneuron	CNS	• Very short axon • Unmyelinated • Cell body on the side of axon	Relay nerve impulses between sensory and motor neurons.
Motor neuron	PNS	• Long axon • Usually myelinated • Cell body at one end.	Conduct impulses from the CNS to effectors.

Nerve pathway

A **nerve pathway** describes the pathway of nerve impulses as they travel between sensory receptors, the CNS, and effectors. The most common effectors in nerve pathways are neurons, muscle cells, and endocrine cells. The stages of a nerve pathway are described below.

1. Sensory receptors detect a stimulus.
2. Sensory neurons transmit the nerve impulse from receptors to the CNS.
3. The CNS interprets the nerve impulse and coordinates the response.
4. Motor neurons transmit the nerve impulse from the CNS to effectors.
5. Effectors facilitate the response to the original stimulus.

Figure 3.19 shows the nerve pathway involved when a person sees a ball moving toward them.

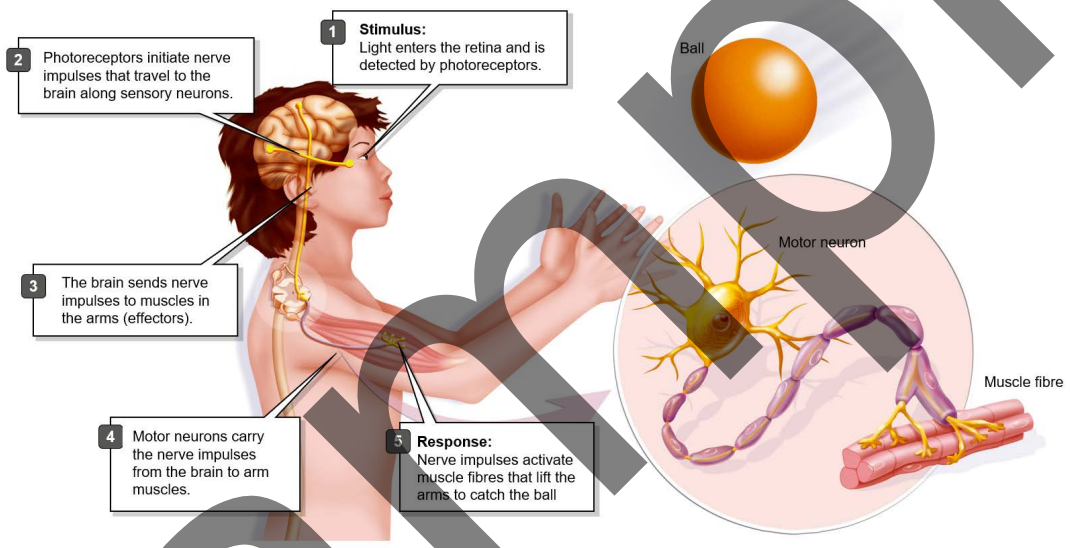


Figure 3.19: Nerve pathway involved in response to stimuli.

Sensory pathways are nerve pathways between receptors in sensory organs and the brain that involve sensory neurons only. Figure 3.20 shows the nerve pathway involved in the sense of taste.

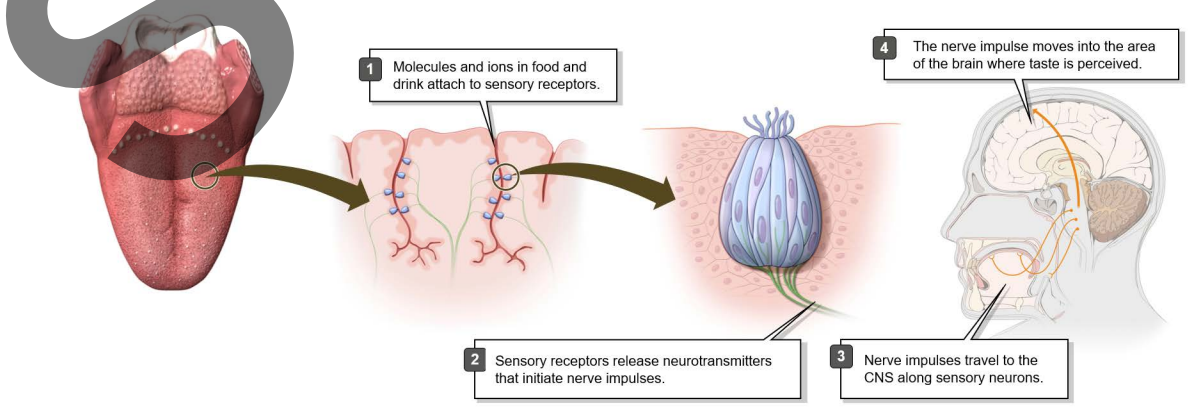


Figure 3.20: Nerve pathway involved in the sense of taste.

Reflex response

A **reflex** is an automatic and involuntary response to a stimulus that is not under the direct control of the brain. A reflex occurs via specialised nerve pathways called **reflex arcs** that allow an organism to respond rapidly to a stimulus. In a simple reflex arc, the stimulus activates sensory receptors that send nerve impulses to the spinal cord. Relay neurons in the spinal cord then transmit the impulse directly to a motor neuron which activates an effector. Figure 3.21 shows the patellar reflex.

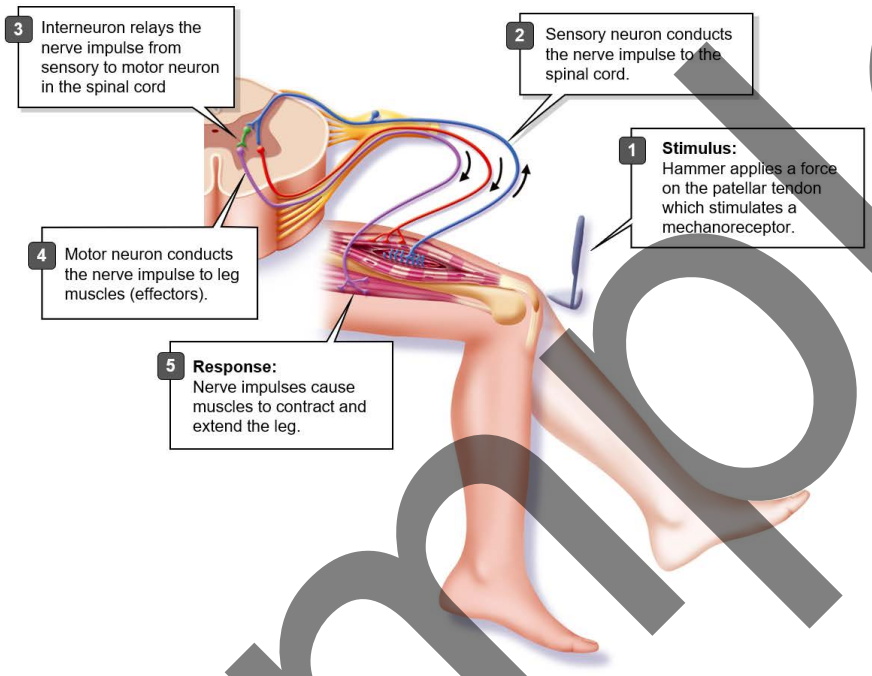


Figure 3.21: The patellar reflex.

A reflex is a rapid response as the nerve impulse is conducted along very few neurons and crosses very few synapses. Reflexes serve a range of vital functions including responding to environmental stimuli, avoiding and reducing tissue damage, maintaining balance and posture, finding food and mates, and escaping from predators. The table below identifies some common reflexes in mammals.

Reflex	Stimulus	Sensory receptor	Response
Pupillary	Change in light intensity	Photoreceptor	Muscles in the iris adjust the diameter of the pupil.
Stretch	Stretching of muscles	Mechanoreceptor	Muscles contract to maintain balance and posture.
Coughing and sneezing	Irritation of airways	Chemoreceptor	Diaphragm and intercostal muscles contract causing violent expiration.
Shivering	Low body temperature	Thermoreceptor	Skeletal muscles shake and generate heat by expending energy.
Nociceptive flexion reflex	Pain	Nociceptor	Muscles contract and withdraw from the painful stimulus.

3.4: The endocrine system

The endocrine system releases hormones that are amino acid derivatives, peptides, proteins, or steroids.

Hormones travel to target sites via the blood.

Hormones can alter the metabolism of target cells, tissues, or organs.

Hormonal responses are stimulated by either the nervous system or other hormonal messages.

- Compare the action of insulin and glucagon in blood sugar regulation.
- Describe how diabetes can result from a hormonal imbalance.
- Describe the role of thyroid-stimulating hormone in the production of thyroxine.
- Describe the action of thyroid-stimulating hormone and thyroxine in metabolism.
- Describe the role of antidiuretic hormone (ADH) in osmoregulation.
- Discuss links between osmoregulation, blood volume, and blood pressure.
- Describe the role of adrenaline in the 'fight or flight' response.

The **endocrine system** regulates growth, development, reproduction and homeostasis in humans and other animals. The endocrine system is composed of glands and organs that produce and secrete chemical messengers called **hormones**. Figure 3.22 shows the major endocrine glands and organs in the human body.

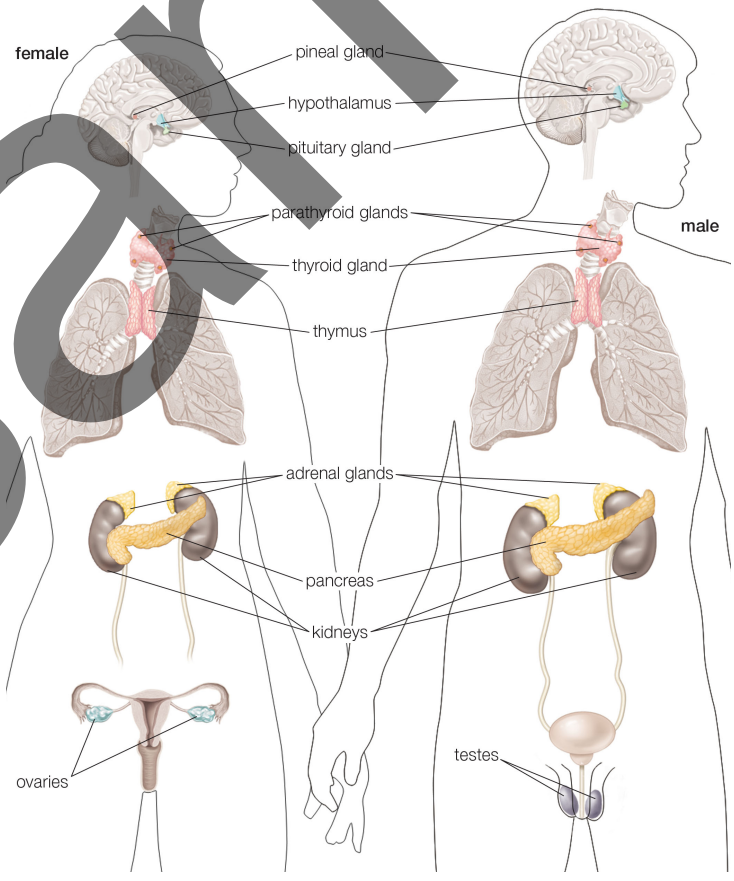


Figure 3.22: The major endocrine organs in the human body.

Hormone pathways

A **hormone pathway** describes the path of a hormone between secretory and target cells in the body. The stages of a hormone pathway are described below.

1. A stimulus causes secretory cells to synthesise the hormone.
2. The hormone is transported from secretory cells to the blood.
3. The hormone is transported in the blood to target cells.
4. The hormone binds to complementary receptors, forming a hormone-receptor complex (HRC) that initiates a signal transduction pathway in the target cell.
5. The signal transduction pathway produces enzymes or proteins that facilitate a response to the original stimulus.

Peptide hormones and most amino acid derivatives are water-soluble and travel freely in blood as they dissolve readily in blood plasma. However, water-soluble hormones cannot diffuse passively across the lipid bilayer and must bind to complementary receptors in the cell membrane of target cells. Steroid hormones and some amino acid derivatives are insoluble in water and become attached to special transport proteins in the blood that increases their solubility in blood plasma. Hormones that are insoluble in water are highly soluble in the lipid bilayer and diffuse passively across the cell membranes and bind to a receptor protein in the cytoplasm or nucleus of target cells. In both cases, the hormone-receptor complex initiates the response, as shown in Figure 3.25.

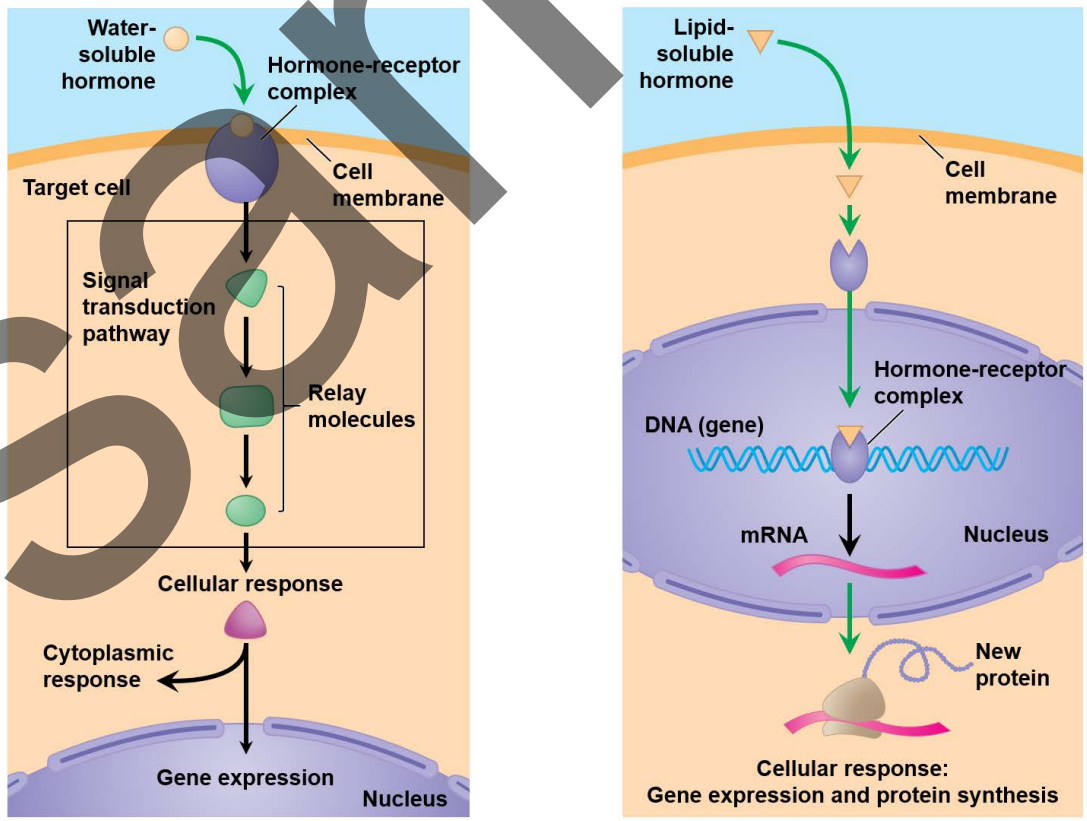


Figure 3.25: Actions of hormones on target cells.

Glucagon and insulin

Glucagon and insulin are peptide hormones produced and secreted by alpha and beta cells in the pancreas in response to changes in blood glucose levels. When levels decline, alpha cells release glucagon, which initiates responses that increase blood glucose. When levels rise, beta cells secrete insulin, which initiates responses that decrease blood glucose. Figure 3.26 summarises the actions of insulin and glucagon in the regulation of blood glucose levels in the human body.

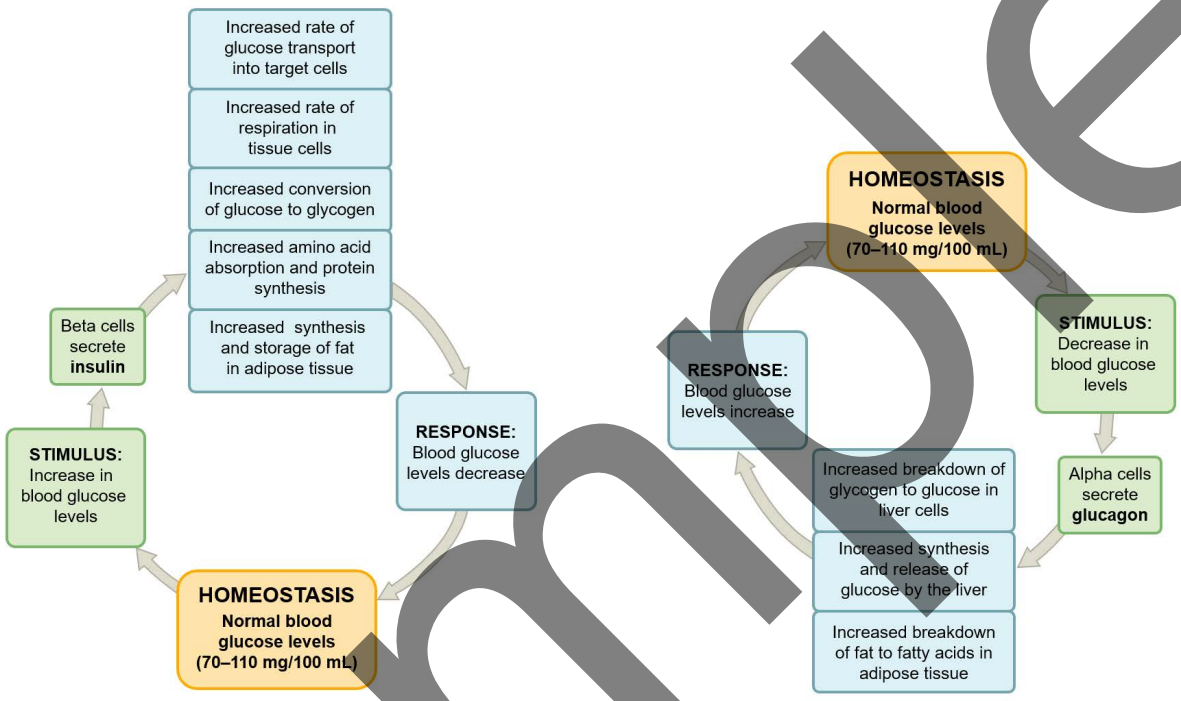


Figure 3.26: Actions of insulin and glucagon.

Animals store glucose primarily in liver and muscle cells as a large molecule called **glycogen**. Insulin binds to liver and muscle cells and initiates the synthesis of glycogen when blood sugar levels are high. Conversely, glucagon binds to liver cells and initiates the breakdown of glycogen to glucose when blood sugar is low. Figure 3.27 shows the structure of glycogen.

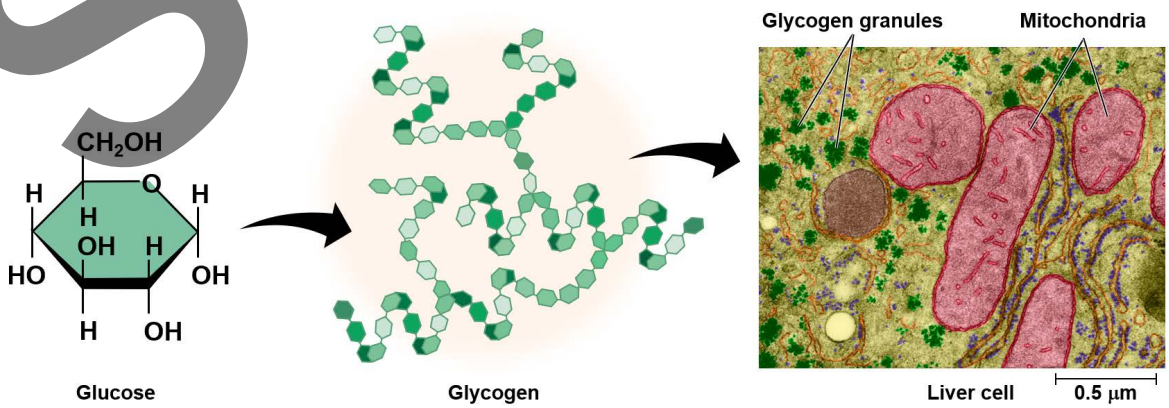


Figure 3.27: The structure of glycogen.

Thyroid hormones

Triiodothyronine (T_3) and **thyroxine** (T_4) are amino acid derivative hormones produced and secreted by follicle cells in the thyroid gland in response to changes in body temperature. When body temperature is below the tolerance limit, thermoreceptors in the hypothalamus detect the change and stimulate neurosecretory cells to secrete **thyrotropin-releasing hormone** (TRH) into the anterior pituitary gland. TRH stimulates secretory cells in the pituitary gland to release **thyroid-stimulating hormone** (TSH) which travels to follicular cells in the thyroid gland. TSH binds to complementary receptors and activates key enzymes that synthesise T_3 and T_4 in follicular cells. T_3 and T_4 travel to target cells where they increase the rate of metabolism and generate heat which increases body temperature and reduces the original stimulus. Figure 3.29 summarises the actions of TRH, TSH, and thyroid hormones in response to low body temperature.

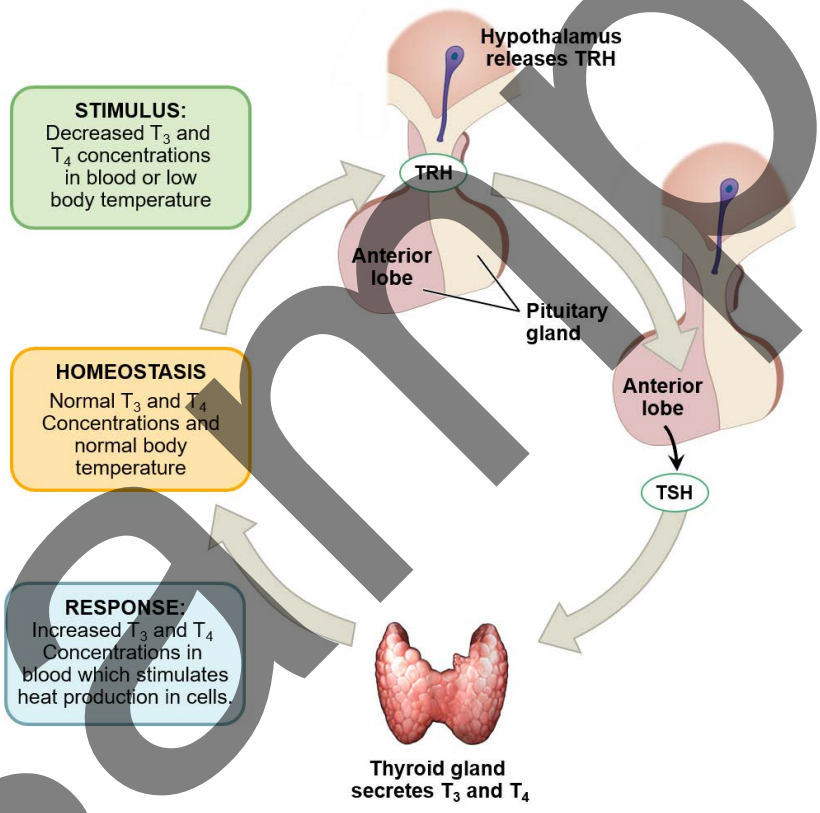


Figure 3.29: Actions of thyroid hormones in response to low body temperature.

An imbalance in thyroid hormones can cause disorders called **hyperthyroidism** and **hypothyroidism**. In hyperthyroidism, too much T_3 and T_4 circulate in the blood which increases the rate of metabolism in target cells resulting in overheating, irritability, high weight loss, high blood pressure, and Graves' disease. In hypothyroidism, too little T_3 and T_4 circulate in the blood, which decreases the rate of metabolism in target cells resulting in low body temperature, lethargy, weight gain, low blood pressure and goitre. One cause of hypothyroidism is a lack of dietary **iodine** which is required for the synthesis of T_3 and T_4 in thyroid follicular cells.

Question 104

Gastric juice is a mixture of chemicals secreted by gastric glands during digestion.

The nervous and endocrine systems stimulate gastric glands in the stomach to secrete gastric juice.

(a) Describe differences in the signalling pathways used by the nervous and endocrine systems.

(2 marks) KA1

(b) The graph shows the volume of gastric juice produced by nervous and hormonal stimulation.

(1) Describe two pieces of evidence that curve A represents nervous stimulation.

(2 marks) KA2

(2) State two stimuli that initiate the nervous stimulation of gastric glands.

(2 marks) KA2

(3) State the evidence that partially-digested food is the stimulus for hormonal secretion.

(1 mark) KA2

First cells

Scientists have observed the ability of vesicles to grow, divide (reproduce), and maintain an internal chemical environment that sustains metabolism. These observations led to the hypothesis that the first simple cells were an assembly of simple organic compounds, macromolecules and genetic material packaged into vesicles with a lipid bilayer (Figure 4.04).

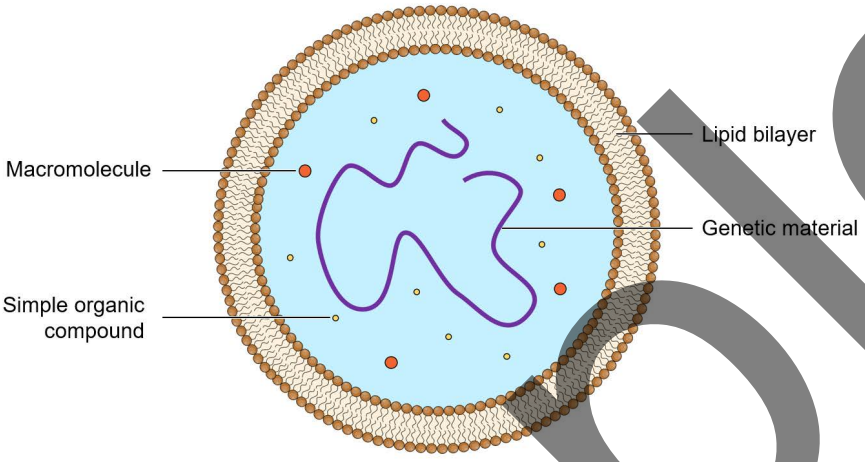


Figure 4.04: Structure of the first simple cells.

Scientists hypothesise that the genetic material in the first simple cells was RNA rather than DNA as short chains of RNA can assemble spontaneously from nucleotides, and mature RNA molecules can act as a template for RNA synthesis as shown in Figure 4.05.

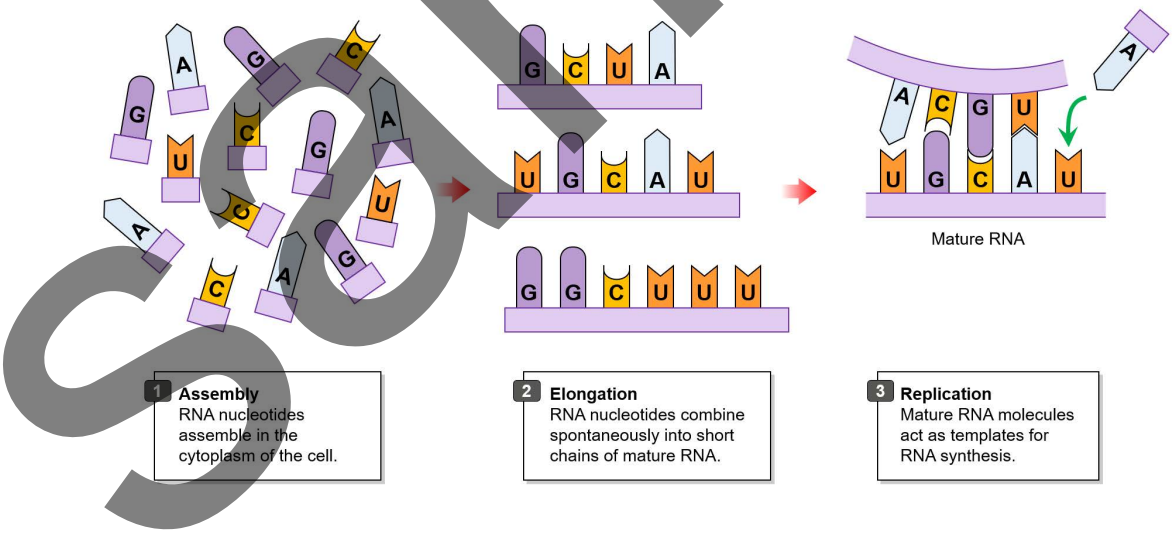


Figure 4.05: Assembly and self-replication of RNA molecules.

In the early 1980s, scientists discovered RNA molecules called **ribozymes** which catalyse many different metabolic reactions including the ability to replicate itself and other RNA molecules. Modern cells contain ribozymes including ribosomal RNA (rRNA) which catalyses the formation of polypeptides from amino acids. The ability of ribozymes to self-replicate and catalyse metabolic reactions is evidence that the genetic material was RNA rather than DNA in the first simple cells.

Endosymbiotic theory

The oldest fossils of eukaryotic cells date back 2.1 billion years. The first eukaryotes were single-celled organisms with membrane-bound organelles including a nucleus, endoplasmic reticulum, mitochondria, and chloroplasts. The **endosymbiotic theory** proposes that the first eukaryotes formed from the endosymbiosis of prokaryotes. The theory states that mitochondria and chloroplasts were formerly small prokaryotes that gained entry to a host cell as undigested prey or internal parasites. Over time, the endosymbionts became interdependent with the host leading to the formation of the first single-celled eukaryotes as shown in Figure 4.08.

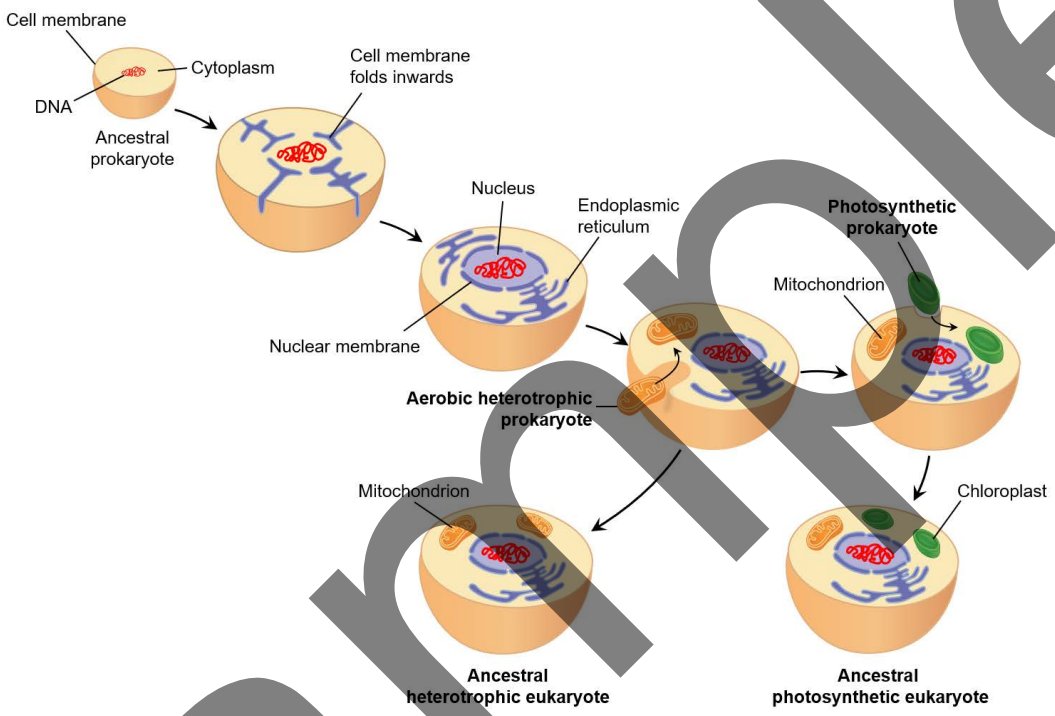


Figure 4.08: Formation of eukaryotic cell from endosymbiosis of prokaryotic cells.

The table below describes supporting evidence for the endosymbiotic theory.

Evidence	Description
Independent replication	Mitochondria and chloroplasts replicate independently of the host cell through binary fission which is the process of cell division in prokaryotes.
DNA	Mitochondria and chloroplasts contain single, circular DNA molecules which are also present in prokaryotes.
Ribosomes	Mitochondria and chloroplasts have their own ribosomes that are a similar size and structure to those in prokaryotes.
Membrane structure	Mitochondria and chloroplasts have a double membrane and the inner membranes are similar to the cell membranes of prokaryotes.
Membrane composition	The membranes of mitochondria and chloroplasts contain proteins and lipids found only in prokaryotes.

DNA-DNA hybridisation

DNA-DNA hybridisation is a technique used to measure the degree of similarity between the DNA of two organisms. DNA extracted from two organisms is mixed and heated, which separates the two polynucleotide strands of the DNA molecules. The mixture is cooled, and the single strands of DNA from the two organisms combine forming hybridised DNA molecules, as shown in Figure 4.12.

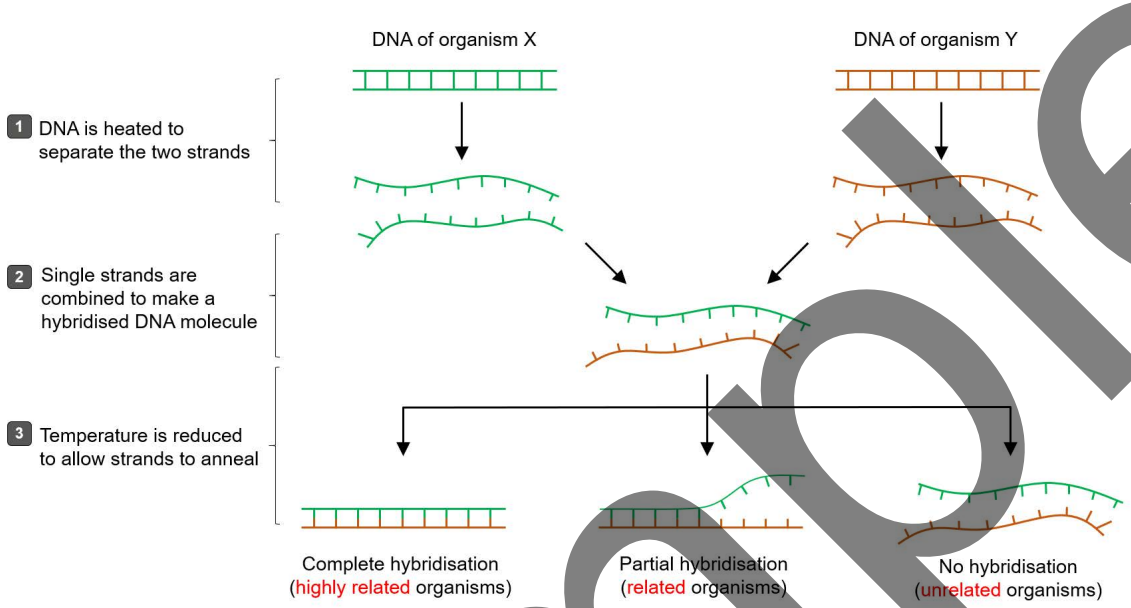


Figure 4.12: DNA-DNA hybridisation technique.

Scientists determine the degree of hybridisation by measuring the temperature at which the hybridised DNA molecules denature. Hybridised molecules that have a high degree of similarity will separate at higher temperatures as more energy is required to overcome the extensive hydrogen bonding between the hybridised polynucleotide strands.

Example 4.03

DNA hybridisation was used to determine evolutionary relationships between different species of bacteria. Samples of hybridised DNA from five species of bacteria were prepared, and the percentage hybridisation of each sample was measured.

Species of bacteria	Percentage hybridisation
A and B	65.3
A and C	91.4
A and D	86.9
A and E	75.4
A and A	100

The data reveals that species A and C are the most closely related given the greater percentage hybridisation, and A and B are the least closely related given the smaller percentage hybridisation.

Question 108

The table below shows the results of DNA-DNA hybridisation experiments between a human gene (*Homo sapiens*) and the same gene in four other primates.

Primate species	Percentage hybridisation
Homo sapiens and *Homo sapiens*	100
Homo sapiens and *Pan troglodytes*	97.6
Homo sapiens and *Gorilla gorilla*	96.5
Homo sapiens and *Hylobates hoolock*	94.6
Homo sapiens and *Macaca mulatta*	91.0

(a) State and explain the primate species most closely related to humans.

(2 marks) **KA2**

(b) The percentage hybridisation was determined by heating the hybridised DNA molecules until the two polynucleotide strands separate.

Explain why this method was used to determine percentage hybridisation.

(2 marks) **KA2**

(c) Haemoglobin is a protein in the red blood cells of primates.

Explain how haemoglobin is used to investigate evolutionary relationships between primates.

(2 marks) **KA2**

(d) Mitochondrial DNA (mtDNA) and genes for ribosomal RNA (rRNA) are used to determine evolutionary relationships between living things.

State why mtDNA and genes for rRNA are used to determine evolutionary relationships between living things.

(1 mark) **KA2**

Phylogenetic tree diagrams represent evolutionary relationships.

Mutations accumulate over time, and the rate of mutation is relatively constant over time which enables its use as a clock.

More closely related species have fewer differences in their DNA sequences and have separated more recently from a common ancestor than distantly related species.

- Draw and analyse simple phylogenetic tree diagrams to represent evolutionary relationships.

A **phylogenetic tree** is a diagram depicting the relationships among groups of living things, the probable evolutionary history of species, and the patterns of descent from recent common ancestors. The branching diagrams reflect the hierarchical classification of living things into taxa, as well as the branching of newly evolved species from a common ancestor. Figure 4.13 shows a phylogenetic tree relating to the classification of organisms into taxa such as order, family, genus and species.

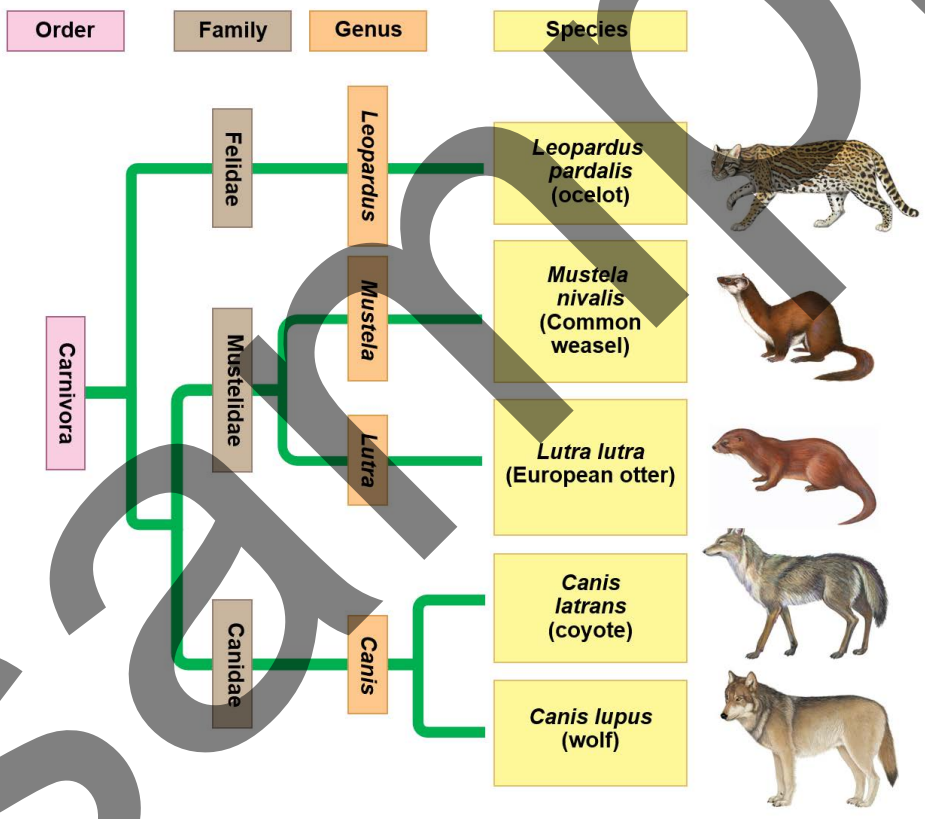


Figure 4.13: Phylogenetic tree showing evolutionary relationships between taxa.

The phylogenetic tree reveals relationships between the five species as well as identifying their most recent common ancestor. The diagram shows that all five species are descended from a common ancestor from the order carnivora. This ancestor gave rise to members of the families; Felidae, Mustelidae, and Canidae. The diagram also shows that the family Mustelidae branched into two separate genera called Mustela and Lutra and that the genus Canus branched into two separate species which includes the coyote and wolf. The diagram shows that the coyote and wolf are the most closely related organisms as they share the most recent common ancestor.

Cladistics

Cladistics is a biological classification system that involves the categorisation of organisms based on shared characteristics. Cladistics groups organisms into **clades** which are groups of species including a common ancestor and all its descendants. The clades are displayed in a type of phylogenetic tree called a **cladogram**. Cladistics is based on the concept proposed by Charles Darwin that organisms share characters with their ancestors but also show new derived characters that distinguish them from their ancestors. There are two main types of characters in cladistics:

1. **Shared ancestral characters**, which are characters that originated in an ancestor of the taxon. These are used to group organisms into clades.
2. **Shared derived characters**, which are characters unique to a particular clade. These distinguish clades and form the branching points of a cladogram.

An important step in cladistics is the identification of **ingroups** and **outgroups**. The ingroup is the species or taxa being studied and the outgroup is a closely-related species that diverged before the ingroup. Comparative genomics is used to compare ingroup species with the outgroup to differentiate between shared ancestral and shared derived characters.

Example 4.04

The cladogram below was constructed using cladistics. The frog represents the outgroup and four other tetrapods represent the ingroup. The presence or absence of traits is indicated as 0 if the trait is absent or 1 if the trait is present. The cladogram is constructed from a series of branch points represented by the emergence of a lineage with a new set of derived traits such as the presence or absence of an amnion, hair, mammary glands and period of gestation (Figure 4.14).

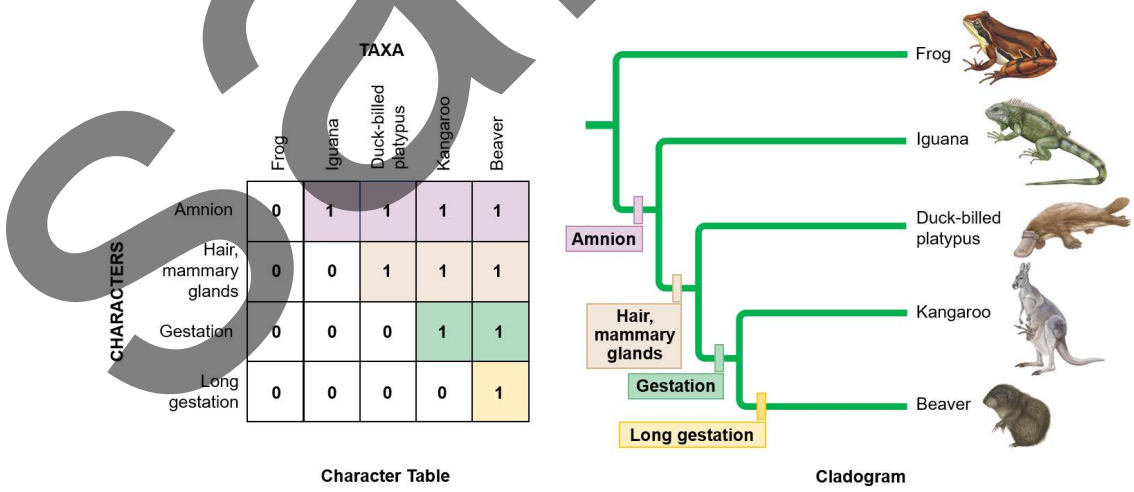


Figure 4.14: Cladogram showing relationships between five animal species.

The phylogenetic tree shows the iguana is the most closely related species to the frog as they share a recent common ancestor and that the beaver is the most distantly related to the frog.

4.3: Defining species

Different criteria are used to define a species depending on the mode of reproduction.

A species that reproduces sexually can be defined by the ability of its members to actually or potentially interbreed to produce fertile offspring.

Other criteria used to define a species include morphological similarity, biochemical similarity and sharing a common gene pool.

Reproductive isolating mechanisms act to maintain distinct species.

- Describe pre-zygotic (prevention of zygote formation) mechanisms including temporal isolation, behavioural isolation, mechanical isolation, and gamete isolation.
- Describe post-zygotic (prevention of fertile hybrids) mechanisms including hybrid inviability and hybrid sterility.

Species is a Latin word meaning "kind" or "appearance". Biologists use different criteria for defining a species. The **morphological species concept** defines a species as a distinct group of organisms with a unique set of morphological characteristics such as body shape and structure. The morphological species concept relies on subjective criteria and is limited because groups that look quite different can be members of the same species and groups that look similar can belong to different species.

Example 4.07

Figure 4.20 shows several members of the species *Helicornis erato,* which are butterflies native to South America. The butterflies have different morphologies but are members of the same species.

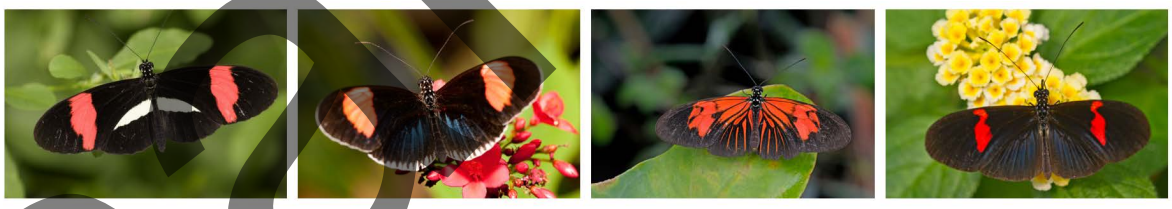
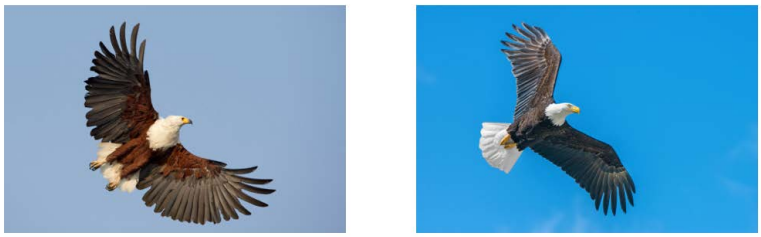


Figure 4.20: members of the same species with different morphologies.

Example 4.08

Figure 4.21 shows members of two different eagle species with similar morphologies.



African fish eagle (*Haliaeetus vocifer*) Bald eagle (*Haliaeetus leucocephalus*)

Figure 4.21: Different species with similar morphologies.

Reproductive isolation

Reproductive isolation is the existence of biological factors that act as barriers preventing two distinct species from interbreeding and producing viable and fertile offspring. Reproductive isolation mechanisms prevent the formation and survival of hybrids and are classified by whether the barrier acts before or after fertilisation. **Pre-zygotic barriers** prevent the formation of a zygote by inhibiting fertilisation, and **post-zygotic barriers** prevent zygotes from developing into healthy and fertile hybrid offspring. The table below identifies and describes several reproductive isolation barriers.

Type of barrier	Description	Effect
Pre-zygotic		
Temporal isolation	Two species breed at different times	Mating is prevented
Behavioural isolation	Two species fail to respond to each other's courtship displays or mating rituals.	Mating is prevented
Mechanical isolation	Two species are physically incapable of mating	Mating is prevented
Geographical isolation	A population becomes separated by a geographic barrier such as mountain ranges.	Mating is prevented
Habitat isolation	Two closely related species rarely encounter each other as they occupy different habitats.	Mating is prevented
Gametic isolation	The gametes of two species fail to fuse or survive in the reproductive tract of the female.	Fertilisation is prevented
Post-zygotic		
Hybrid inviability	Hybrid fails to develop after fertilisation	No offspring produced
Hybrid sterility	Hybrid reaches sexual maturity but is infertile and cannot reproduce.	No offspring produced

Example 4.09

Figure 4.24 shows two skunks that are classified as separate species due to reproductive isolation. The Eastern spotted skunk breeds in late summer and the western spotted skunk breeds in late winter which is an example of temporal isolation.

Eastern spotted skunk (*Spilogale putorius*) Western spotted skunk (*Spilogale gracilis*)

Figure 4.24: Skunk species that are reproductively isolated.

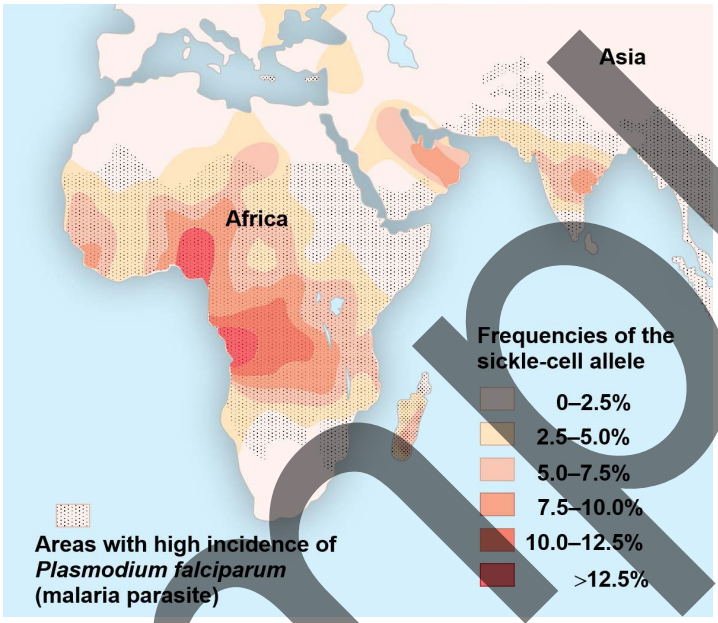
Question 118

Malaria is an infectious disease caused by a parasite that infects red blood cells.

Sickle-cell anaemia (SCA) is an inherited disorder that confers resistance to malaria by distorting the shape of red blood cells, which reduces the ability of the parasite to cause infection in humans.

Individuals with one copy of the sickle-cell allele are relatively resistant to malaria.

The map below shows the frequencies of the sickle-cell allele in areas with a high instance of malaria.



SCA is caused by a mutation in the β-globin gene.

(a) State one reason why mutations rarely change allele frequencies in human populations.

_____

_____ (1 mark) KA2

(b) Describe and explain the data in the diagram above using the concept of natural selection.

_____

_____

_____

_____

_____

_____ (3 marks) KA2

(c) Mutation is one source of genetic variation in the human population.

State one other source of genetic variation in humans.

_____ (1 mark) KA2

4.5: Speciation

Speciation may result from an accumulation of genetic changes influenced by different selection pressures or genetic drift in geographically isolated populations.

- Describe the process of speciation due to physical separation (allopatric speciation).
- Compare allopatric and sympatric speciation.

Biologists estimate between 10 and 100 million species live on Earth, of which only 1.8 million have been named and described. Evidence shows that all living and extinct species descended from a common ancestor that lived on Earth around 3.5 billion years ago. Factors such as mutation, sexual reproduction, natural selection and genetic drift have caused cumulative changes in the gene pools of populations over time which has resulted in the evolution of new and distinct species.

Speciation

Speciation is the emergence of a new species and occurs when gene flow has effectively stopped between populations due to reproductive isolation. Reproductive isolation mechanisms prevent gene flow between the gene pools of two populations such that changes in the allele frequency in one population are not shared with the other. Cumulative changes in the allele frequencies of the two distinct gene pools result in members of the two populations being unable to interbreed and produce viable and fertile offspring. Two mechanisms that facilitate the emergence of a new species are called **allopatric speciation** and **sympatric speciation** (Figure 4.42).

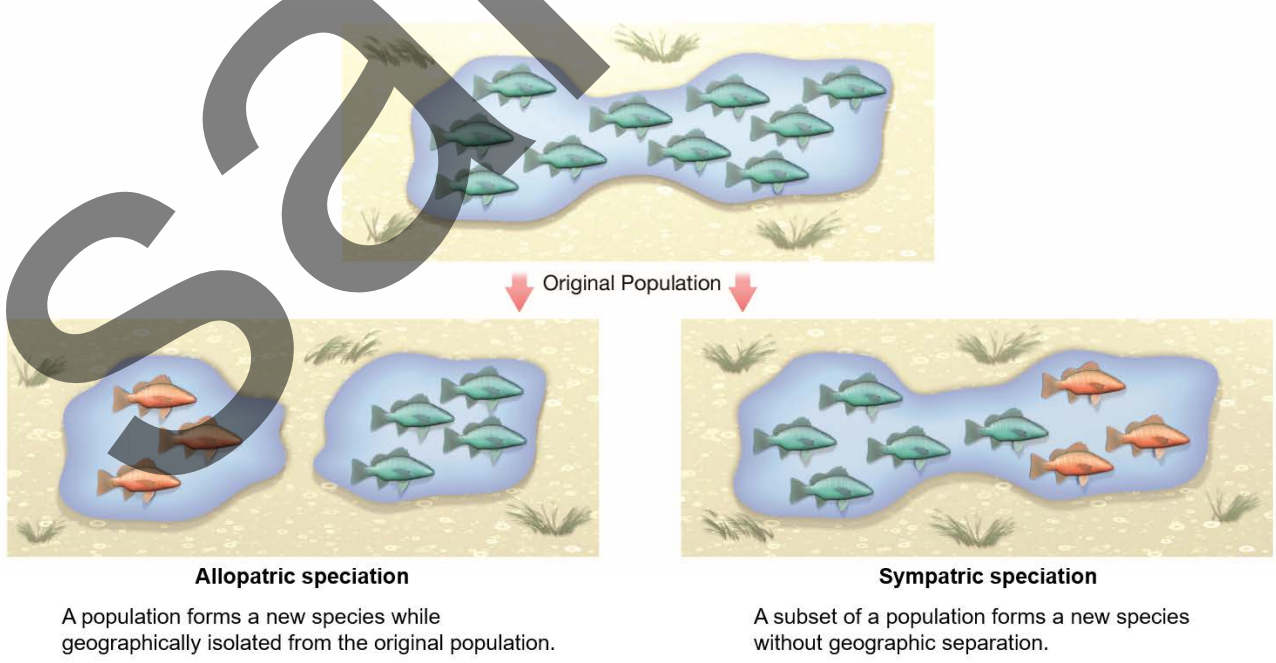


Figure 4.42: Mechanisms of speciation.

Sympatric speciation

In sympatric speciation, speciation takes place in geographically overlapping populations when a subset of a population forms a new species without geographic separation. Sympatric speciation is the result of reproductive isolation in the absence of any physical barrier that prevents gene flow between two populations and may result from polyploidy, natural selection, or sexual selection. Sympatric speciation is also caused by temporal, habitat, gametic and behavioural isolation.

Example 4.23

Polyploidy is the presence of extra sets of chromosomes due to accidents during cell division and is common in plants but not animals. Polyploidy leads to the emergence of an **allopolyploid** which is a species with multiple sets of chromosomes derived from different species, as shown in Figure 4.44.

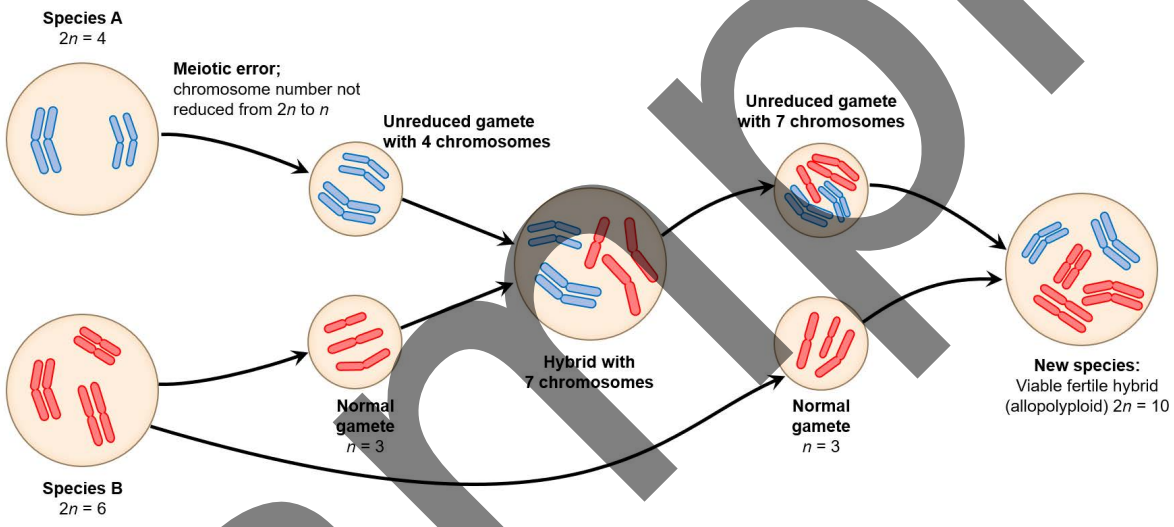


Figure 4.44: Formation of an allopolyploid.

Rapid changes in chromosome numbers cause sympatric speciation in plants that share a habitat. Polyploidy forms new plant species in a single generation as populations that differ in chromosome number are reproductively isolated. Figure 4.45 shows two strawberry species that share a habitat but are reproductively isolated due to the effect of sympatric speciation caused by polyploidy.

Figure 4.45: Reproductively isolated strawberries.

Example 4.24

Sympatric speciation can also result from the appearance of new ecological niches. Figure 4.46 is an illustration showing differences in the feeding behaviour and morphology of four species of cichlid fishes that inhabit Lake Victoria in Eastern Africa. The four species are descended from a common ancestor, but each population has diverged due to the acquisition of separate ecological niches. Natural selection altered the allele frequencies of the ancestral populations over time by selecting for alleles that increased reproductive success in their niche. Eventually, natural selection led to sympatric speciation as the four populations became reproductively isolated.

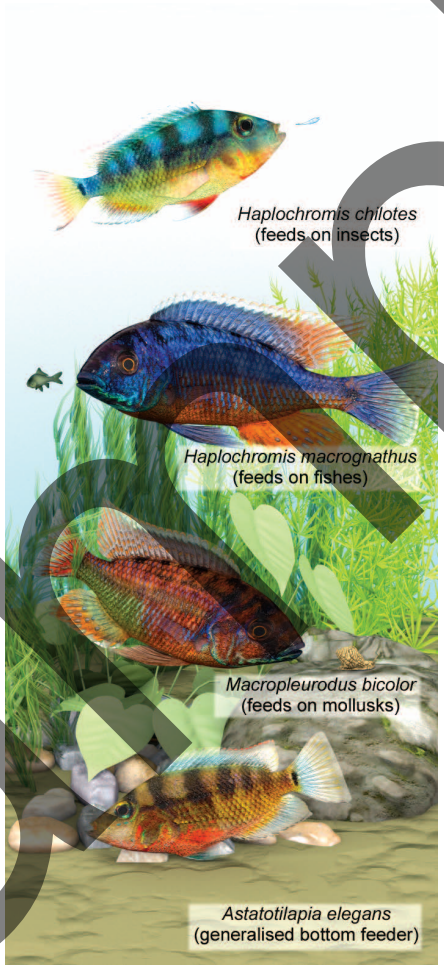


Figure 4.46: Sympatric speciation in cichlid fishes.

Sympatric speciation can also result from **sexual selection** in which members of a species favour certain characteristics in individuals of the other sex. Sexual selection for mates of different colours has likely contributed to sympatric speciation in Lake Victoria which is home to many distinct species of cichlid fish that vary in colour from blue to red. Cichlid fish species that inhabit shallow water are blue and prefer to mate with other blue-coloured fish, whereas cichlid species that live in deeper water are red and prefer to mate with other red-coloured fish. All species evolved from a common ancestor in the same habitat which is evidence that sexual selection has led to sympatric speciation in cichlid populations within Lake Victoria.

4.6: Types of Evolution and Succession

Similar selection pressures in different environments may lead to convergent evolution.

When new niches become available to a species, as a result of succession or environmental change, different selection pressures may lead to divergent evolution or adaptive radiation.

- Recognise and give examples of convergent evolution.
- Recognise and give examples of adaptive radiation.
- Describe the process of succession.

Natural selection does not create new traits but edits or selects for favourable alleles already present in the gene pool of a population. The local environment and selection pressures determine the alleles that are selected for in any population with alleles for advantageous traits being more likely to increase in frequency over time.

Convergent evolution

Convergent evolution is the process whereby natural selection causes distantly related populations to independently evolve similar structural, biochemical or behavioural characteristics due to similar selection pressures in their respective environments. Characteristics that result from convergent evolution are called **analogous characteristics**.

Example 4.25

Cacti in North America and euphorbs in North Africa have evolved analogous characteristics including smaller leaves, short spines, succulent stems, a fibrous root system, and crassulacean acid metabolism as a result of similar selection pressures in their desert environments. Similarly, the flying squirrel in North America and sugar glider in Australia have independently evolved wing-like flaps for leaping from trees, and big eyes for foraging at night (Figure 4.47).

Figure 4.47: Examples of convergent evolution in plants and animals.

Divergent evolution

Divergent evolution is the process of developing two or more species from a common ancestor over time. Divergent evolution is a slow process and occurs when two populations become separated by a geographic barrier and experience different selective pressures. Under certain conditions, a phenomenon known as **adaptive radiation** may occur in which an ancestral population experiences multiple speciation events in a relatively short period. Adaptive radiation occurs when a range of new ecological niches become available such as the formation of new islands or through mass extinctions. In such cases, multiple speciation events occur as many populations diverge from a common ancestor over a short period, and each experiences different selection pressures leading to changes in the allele frequencies of its gene pool.

Example 4.26

Around 200 million years ago, mammals were small, nocturnal animals that lived in tall trees and underground burrows. Around 65 million years ago, mammals spread out and rapidly occupied the ecological niches vacated by dinosaurs following their extinction. Adaptive radiation resulted in the rapid speciation of mammals adapted to running, leaping, climbing, swimming, and flying. Evidence shows that adaptive radiation led to more than 5 000 speciation events in mammals over 65 million years. Figure 4.48 shows several mammals that evolved from a recent common ancestor.

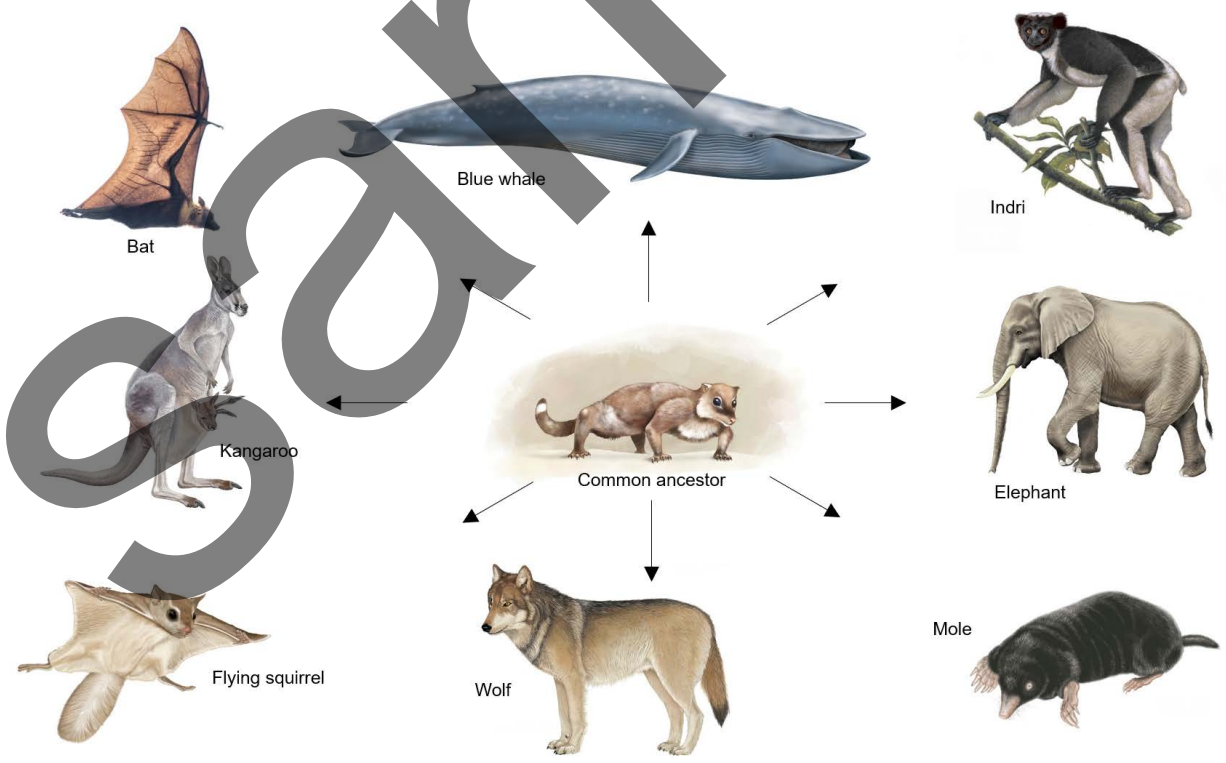


Figure 4.48: Adaptive radiation in mammals.

Solutions: Chapter One			
Question	Part	Author's response	Marks
1	(a)	A: phosphate group	1
		B: sugar	1
		C: base/nucleobase/nucleotide base/nitrogenous base	1
		D: hydrogen bond	1
	(b)	Thymine = 31.2%	1
		Cytosine = 18.8%	1
		Guanine = 18.8%	1
	(c)	**Any one similarity** Both are composed of nucleotides; Both contain adenine, cytosine, and guanine; Both contain a sugar; Both contain phosphate;	1
		Any one difference Sugar in DNA is deoxyribose/sugar in RNA is ribose; DNA is double-stranded/RNA is single-stranded; DNA contains thymine/RNA contains uracil; DNA is a much longer molecule.	1
2	(a)	J: Adenine	1
		K: Cytosine	1
		L: Thymine	1
	(b)	Free DNA nucleotides bind to exposed nucleotides on the exposed strands of the parent DNA molecule via complementary base pairing;	1
		Complementary base pairs interact through hydrogen bonds.	1
	(c)	Helicase breaks the hydrogen bonds between the bases on the polynucleotide strands of the parent DNA molecule.	1
		DNA polymerase joins the sugar and phosphate groups of adjacent nucleotides creating the sugar-phosphate backbone of the newly synthesised polynucleotide strands.	1
3	(a)	The conservative model states that the parent DNA molecule serves as the complete template for a DNA molecule made from two new strands.	1
		The semiconservative model states that the two strands of the parent DNA molecule separate, and each serves as a template for the synthesis of a new DNA strand.	1
		The dispersive model states that the original DNA molecule breaks into fragments that serve as templates for the synthesis of new DNA fragments.	1
	(b)	Semi-conservative model.	1

Solutions: Review Test 2			
Question	**Part**	**Author's response**	**Marks**
1	(a)	K	1
	(b)	J	1
	(c)	M	1
	(d)	J	1
	(e)	L	1
	(f)	L	1
	(g)	K	1
	(h)	L	1
	(i)	J	1
	(j)	L	1
2	(a)	**Any one:**	1
		Offspring are produced without the need for a partnership; It occurs over a short period/is more rapid than sexual reproduction; The energy requirements for reproduction are minimal; It can occur in various environments; Favourable genes are guaranteed to be passed to the next generation; Maturity is rapid;	
	(b) (1)	$6CO_2 + 6H_2O \xrightarrow[\text{chlorophyll}]{\text{light}} C_6H_{12}O_6 + 6O_2$	2
	(b) (2)	Autotroph	1
	(c) (1)	Aerobic respiration	1
	(c) (2)	Adenosine triphosphate/ATP;	1
	(d)	In prophase, the nuclear membrane is broken down, and the replicated chromosomes condense and become visible under a light microscope; In metaphase, the replicated chromosomes are arranged along the equator of the parent cell by spindle fibres.	1 1
	(e) (1)	Asexual reproduction does not introduce any significant genetic variation; A lack of genetic variation makes a population more susceptible to infectious disease because very few individuals, if any, possess alleles that combat disease.	1 1
	(e) (2)	The growth of *Phytophthora infestans* on leaves reduces the surface area for the absorption of sunlight; The rate of photosynthesis and aerobic respiration are reduced, which reduces the available energy for growth.	1 1
	(f) (1)	Nucleus	1
	(f) (2)	**Advantage:**	1
		Increases crop yields; Increases profits; Reduces crop waste.	
		Disadvantages:	1
		Metalaxyl can be toxic to birds, fish, beneficial insects, and non-target plants; Metalaxyl can contaminate soil, water, and other vegetation	