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# **CELL BIOLOGY**



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# 1.1 Cell theory









7 Functions of life

Nutrition

Metabolism

Excretion

Response

Growth

Homeostasis

Reproduction

- 1. All organisms are composed of cells: microscopic examination of many organisms has shown that they are all composed of cells; unicellular organisms are still composed of one cell that performs all functions of life.
- 2. Cell is the basic unit of life: cell as a whole can perform the functions of life, while its individual components cannot.
- 3. All cells originate from a pre-existing cell: spontaneous generation of cells is not possible; a cell needs to divide to create another cell.

Even though most organisms fit well into the first two points of the cell theory (A and B), some organisms and tissues seem to contradict it. Muscle fibres are fused, elongated cells with multiple nuclei and as such differ from the common definition of a cell (C). Similarly, fungal hyphae often don't contain dividing walls and are made up multiple fused thread-like cells (D).



If these are hard to remember, think about the following:

Every living being has to eat<sup>①</sup>, process the food it has eaten<sup>②</sup> and excrete the waste<sup>③</sup>, take in the signals from the environment and respond to them<sup>④</sup>, accordingly make sure that it is well balanced<sup>⑤</sup> (food-wise, heat wise, etc.) and finally, use all that to grow<sup>⑥</sup> and pass on its genes<sup>⑦</sup>.



## 1.1.1 Sizes of cells

#### Surface area to volume ratio

Surface: the rate at which the cell can take in, or send out food/waste materials.

Volume: the amount of food a cell needs, or the amount of excretions the cell produces.

Cell growth is limited by two features of the cell: *surface area* and *volume*.

When the cell volume increases, the surface area increases comparably less. This limits the size of a cell because:

- the cell must be able to transport enough food/waste through the surface
- compared to the food needs/excrement production, which is determined by the cell volume.

Example surface area (SA) and volume (V) calculation



Imagine a cell as a cube with sides of length *a*. The surface area (*SA*) can be calculated by adding the SAs of the six faces of the cube and the volume (*V*) by multiplying the sides.  $SA = 6 \times a^2$  and  $V = a^3$ 

Now, compare the growth of cells, starting with a = 1, a = 5 and a = 10.

	size of 1	size of 5	size of 10
side= $a$	a=1	a=5	a = 10
$SA = 6 \times a^2$	SA = 1	<i>SA</i> =150	SA = 600
$V = a^3$	V = 1	V = 125	V=1000

Up to a certain size the SA still exceeds the V, and the cell would be able to import and export enough materials to sustain its life. But as the cell grows bigger the volume will exceed the SA, at which point the cell cannot transport enough materials in and out of the cell to keep up with its food needs/waste production.



## **Typical cell sizes**

Cells have different sizes, from one organism to another as well as within an organism. This difference naturally arises from the difference in cells functions, and needs. The following scheme should help you compare the sizes of different cells:

molecules	cell membrane	virus	prokaryotes	organelles	eukaryotes
1 nm	10 nm	100 nm	1 µm	10 µm	100 µm

#### **Calculating magnification**



 Real cell size

 Exam question: Image of a cell, measurable by a ruler. Magnification in the corner.

 Calculate the real size of the cell in the picture

 Solution:

 Image of the cell = real cell size × magnification

 therefore

Real cell size =  $\frac{\text{Image of the cell}}{\text{magnification}}$ 

Numbers worked out.

#### Microscopes

An electron microscope has a far greater resolving power than a conventional light microscope, meaning an electron microscope can be used to create images of smaller objects with greater resolution.

The limit of resolution is determined by the wavelength of the incident light/electrons. And since the wavelength of electrons is much smaller than that of visible light, they can be used to view objects much smaller and in much more detail.



# **1.1.2 Cell properties**

In multicellular organisms, each cell has its own function and cooperates with other cells to form an organism.

#### **Emergent properties**

Special properties of organisms that arise from complex cell interactions. The function of the whole organism is greater than the sum of individual cell functions.

#### **Cell differentiation**

Specialization of cell function: each cell in a multicellular organism has its own function, which is defined by the expressed genes in that cell. Although all cells possess the same genes, expressed genes are the ones that are "switched on" while all other genes are "switched off".

#### **Stem Cells**

Example.

Example.

Undifferentiated cells that can divide and have the capacity to express (turn on and off) any set of genes, thus able to acquire any function. Therapeutic sources of stem cells include umbilical cord blood, bone marrow and human embryonic stem cells

#### 1.1.3 Examples of stem cell use

#### Stadgardt's disease

Stadgardt's disease is a degenerative disease of the eye (retinal cells) leading to blindness. Human embryonic stem cells are obtained from unsuccessful in vitro fertilizations. These cells are differentiated in the lab towards retinal cells and injected into the eye of patients. The new cells replace the degenerate cells in the retina and restore vision

#### Leukaemia

Leukaemia is the cancer of white blood cells (immune cells). Human cord blood is collected after childbirth. The cord blood contains stem cells that differentiate into white blood cells. A patient with leukaemia is irradiated and given chemotherapy to kill all cancerous white blood cells. The killed cells are then replaced by the matching cord blood cells which are able to differentiate into all kinds of white blood cells in the patient



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# **1.2 Cells and membrane transport**

# 1.2.1 Eukaryotic and prokaryotic cells

The main distinction between the eukaryotic and prokaryotic cells relates to their size and complexity. Prokaryotes, also known as bacteria, are unicellular organisms with a simple, non-compartmentalized structure. Eukaryotes, which can be unicellular or multicellular organisms, are usually bigger with a complex organelle based function.

#### **Prokaryotic cells**



Figure 1.1: Prokaryotic cell

Table 1.1: Functions of prokaryotic cells			
Structure	Function		
Capsule	Protection		
Cell wall	Protection and pressure maintenance		
Cell membrane	Transport of materials		
Cytoplasm	Contains enzymes, food, medium for cellular processes		
Ribosomes	Protein synthesis		
Nucleoid	DNA containing area not enclosed by a membrane		
Plasmid	Extra genetic material (e.g. antibiotic resistance genes)		
Pilli	Communication, DNA exchange, attachment		
Flagellum	Movement		



Make sure you can draw and label this yourself!



# **Eukaryotic cells**



Make sure you can draw and label this yourself!

There are two types of eukaryotic cells: pancreatic cell (animal) and mesophyll cell (plant).

Table 1.2:	Comparison	of animal	cell and	plant cell
1 abit 1.2.	Comparison	or anninai	cen anu	plant con

Structure	Function	Animal cell	Plant cell
Ribosome	Protein synthesis	1	1
Rough endoplasmic reticulum	Protein modifications	1	1
Golgi apparatus	Protein packaging	1	1
Mitochondrion	Site of cell respiration	$\checkmark$	$\checkmark$
Nucleus	Contains chromosomes (DNA)	$\checkmark$	$\checkmark$
Lysosome	Degradation enzyme storage	$\checkmark$	1
Centrioles	Chromosome separation during mitosis	$\checkmark$	1
Vacuole	Food and water storage	×	1
Cell Wall	Maintenance of cell pressure	×	$\checkmark$
Chloroplast	Site of Photosynthesis (food production)	×	1



#### Comparison of Eukaryotic cells and prokaryotic cells

Prokaryotic Cell	Eukaryotic Cell
Naked DNA	DNA wrapped around proteins
DNA in cytoplasm	DNA enclosed by a nuclear envelope
DNA circular	DNA linear
No membrane bound structures	Membrane bound structures such as
	mitochondria, ER, Golgi apparatus
	present which compartmentalize
	functions
Plasmids present	No plasmids
Mitochondria not present	Mitochondria always present
Ribosomes smaller (70S)	Ribosomes larger (80S)

Table 1.3: Comparison of prokaryotic cells and eukaryotic cells

Besides in their structure, the two types of cells also differ in their mode of division. Eukaryotic cells divide by **mitosis** (discussed later) while prokaryotic cells divide by **binary fision**.

## 1.2.2 Cell membrane



Notice the head / tail structure of thephospholipid. In order to isolate the water from the tail areas, phospholipids form a bilayer, thereby exposing the heads to the water instead.

Due to these interactions, the plasma membrane is very stable but is said to be **fluid.** This means that the tails will always be facing tails, and the heads will always face outside, but the position of individual phospholipids in a layer may change.

This property of the membrane also allows it to hosts a variety of other molecules, like proteins and cholesterol. This makes it look like a **mosaic**.



Membrane proteins fulfil various functions:



Figure 1.3: Phospholipid molecules form a phospholipid bilayer, which together with proteins and cholesterol forms cell membranes.

Cholesterol keeps the fluidity of the membrane constant at a variety of temperatures.

Channel Recognition Enzymatic Carrier Receptor • 

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Figure 1.4: Phospholipid molecules form a phospholipid bilayer, which together with proteins and cholesterol forms cell membranes.



# **1.2.3 Membrane transport**

Recall that the main function of the plasma membrane is transport. Generally, transport is defined as passive or active.

Active transport movement of molecules from an area of lower concentration to an area of higher concentration, with the use of energy (against the concentration gradient)
Passive transport movement of molecules from an area of higher concentration to an area of lower concentration (down the concentration gradient)
Passive transport is further divided into two types of diffusion, simple and facilitated.

Simple diffusion passive transport of molecules through a membrane, without the need of protein channels (oxygen diffusion)

**Facilitated diffusion** passive transport of molecules **facilitated** by channel proteins (sodium transport, calcium transport)

Osmosis is a form of passive transport that only refers to the movement of water. The water, like other particles in passive transport, moves from the area where there is more of it, to the area where there is less of it. However, osmosis is defined in terms of the concentration of dissolved molecules:

**Osmosis** movement of water from the area of low solute concentration to the area of high solute concentration.

Active transport, like (passive) facilitated diffusion requires proteins. However, these proteins use energy in form of ATP to pump molecules against their concentration gradient.



Sodium-potassium pump is such a protein, and can be found in many cells including neurons. This pump is described in more detail in the "Human physiology" chapter, but now consider the following points:

- sodium potassium pump is an integral protein that uses ATP to transport molecules across a membrane;
- it transports sodium out of the cell, and potassium into the cell;
- it works against the concentration gradients of both sodium and potassium;
- for every three sodium molecules it transports out, two potassium molecules are transported in.

You should consider two more types of active transport that involve vesicle transport, rather than protein pumps.



**Endocytosis** transport of molecules into the cell through invagination of the plasma membrane and formation of the phospholipid vesicle containing the molecule.

# **1.2.4 Osmolarity**

Imagine a potato cube in a water bath. Mind you that the potato has a much smaller volume compared to that of the water tank.

#### Hypotonic





The ratio of solutes to water inside the potato is much higher than that same ratio in a water bath.

The bath is *hypotonic* compared to the potato.

The water moves from the bath into the potato, making the potato swell.

The ratio of water to solute in the potato is the same as the one in the bath.

Recall that in osmosis, the water moves from where there is more of it to where there is less of it



#### **Hypertonic**



The water bath is more saturated with its solute compared to the potato.

The bath is a *hypertonic* solution compared to the potato.

The water inside the potato will pass into the water bath, trying to dilute it to the same concentration as in the potato.

The potato will shrink, its ratio of solute to water will increase and the osmolarities will be balanced.

# **1.3 Origin of cells**

## 1.3.1 Pasteur's soup

A prior belief was that cells could spontaneously arise from the assembly of inorganic matter. However, Louis Pasteur disputed the belief of spontaneous formation of life in the 19<sup>th</sup> century.

In his simple experiment, he filled two flasks with nourishing soup and then sterilized them. One flask had a straight open neck, while the other had a curved opened neck. Within a week, the straight-necked soup was spoiled and the curved-necked soup was as good as it was on the first day.



The germs found in the spoiled soup, could be found at the entrance of the curved necked, where they got stuck. Therefore, the mould, fungi and bacteria were able to enter the soup from the environment, but were not able to assemble from thin air in the sealed container.



# **1.3.2 Formation of organic molecules**

In order to form cells, first we have to form (relatively) complex molecules. The Miller-Urey experiment showed that:

- the water vapour, ammonia and methane, all found in the early atmosphere, could have spontaneously assembled into amino acids and carbon compounds, in the presence of electricity (lightning);
- if some of the compounds formed at that time on earth were phospholipids, they would have naturally assembled into bilayers, forming early membranes;
- the formation of nucleic acids such as RNA would have given rise to early enzymatic activities, protein assembly and the first genetic information.

# **1.3.3 Endosymbiotic theory**

Next, this theory assumes that more complex eukaryotic cells have evolved from the prokaryotic cells through a symbiotic process.

- Symbiosis is a mutually favourable coexistence of two organisms.
- The existence of genetic material within mitochondria and cholorplasts suggests that these organelles could have been cells of their own.
- The theory suggests that a larger anaerobic prokaryotic cell could have engulfed a smaller aerobic cell, and started coexisting with it.
- The large cell was supplying the smaller one with food, while the smaller cell was converting the food into energy for the larger cell → symbiosis.



# **1.4 Cell division**



The function of mitosis is to create a daughter cell with the identical genome to the mother cell. The process involves replication (=duplication) of DNA (all chromosomes). In order for separation of duplicated DNA to work, the DNA (normally a very long molecule) needs to *supercoil*. Replication is said to be proofread and checked for errors by the cell's machinery.

Remember that mitosis occurs only in eukaryotic cells, while prokaryotic cells divide by binary fision.

Cytokinesis is the division of the cell's cytoplasm and organelles that directly follows mitosis

In plant and animal cells, the process of cytokinesis differs. In plant cell, the kinesis forms through formation of vesicle along the equator of the cells leading to their eventual fusion and formation of the plasma membrane. The vesicle also bring cellulose to form the cell wall around the newly formed plasma membrane. In animal cells, the division of cytoplasm is a result of invagination of the plasma membrane. Actin and myosin are the contractile fibres that create this invagination called cleavage furrow.



# 1.4.1 Cell cycle

From its formation, until division, each cell goes through several phases of the life cycle.

- G1 is the phase in which cells spend the majority of their lifespan: this is the period of growth and performance of its daily functions.
- S is the phase that occurs once the cell has decided to undergo mitosis: this is the period of DNA synthesis (replication)
- G2 is the phase where the cell does its last preparations for mitosis: during G2, the cell duplicates its organelles and prepares enzymes and proteins needed for mitosis



- Cyclins comprise the cell cycle checkpoints
- The first cell cycle check point occurs between G1 and S phase
- Another checkpoint occurs during S phase before the beginning of DNA replication
- If the cyclins are not produced or activated, the cell cannot pass a cell cycle checkpoint

Despite this tightly regulated cell cycle system, some cells manage to escape the checkpoints and form tumours.

**Cancer** is the result of uncontrollable cell division and tumours are the aggregates of cancerous cells.

- Mutagens are agents that cause mutations in the DNA.
- Some of these mutations can be missed by proofreading machinery leading to gene mutations
- UV light is a known mutagen that cause high rate of DNA mutations that can often not be repaired



#### CELL BIOLOGY | Cell division

- Oncogenes are genes of each cell that are responsible for normal cell division
- If these genes are mutated, they often lead to cancer
- Proto-oncogenes are oncogenes that in their mutated state become overactivated and promote cell division leading to tumour formation
- Tumour suppressor genes are genes that negatively regulate the cell cycle, so when mutated, they fail to prevent uncontrollable cell divisions
- Metastasis refers to the movement of the primary cancerous cells to a new formation where they continue to form tumours.

## 1.4.2 Phases of mitosis

Mitosis consists of 4 phases that can be distinguished under the microscope. Due to supercoloiling of the DNA, the chromosomes become visible and can be tracked during these phases.

#### **Prophase**

Figure 1.5: Prophase: nuclear envelope is fractured, chromosomes are becoming thick, centrioles are located and the poles of the cell.



- DNA supercoils, chromosomes condense and become visible.
- Nuclear envelope breaks down.
- Spindle microtubules start forming at the poles of the cell.
- The cell contains double the DNA compared to its G1 phase, the same number of chromosomes.



#### Metaphase

Figure 1.6: Metaphase: chromosomes located at the equator of the cell, each X representing a chromosome, and they're ordered in one single row. Spindle fibres origninate at the poles and attach to centres of X-es each pole, with one chromosome having one spindle from each pole.



- Chromosomes align at the equator of the cell.
- Spindle microtubules attach to the centromeres (centres of the chromosome).

#### Anaphase

Figure 1.7: Anaphase: fibres are shortening towards the poles, and dragging one leg of the X towards the pole. Equal number of chromosome legs are moving to each pole.



- Sister chromatids (legs of each chromosomes, containing identical copies of DNA) are pulled to opposite poles by spindle microtubules.
- Now there is an equal number of chromosomes (DNA molecules) at each pole, but overall, the cell now has double the number of chromosomes compared to prophase.



#### **Telophase**

Figure 1.8: Telophase: two nuclei beginning to form at each pole, and the chromosomes uncoiling and becoming longer.



- Chromosomes begin to uncoil as the nuclear envelope reforms around them.
- The cell contains two identical nuclei and awaits the division of cytoplasm and organelles (= Cytokinesis).

When observing a tissue or a group of cells under a microscope, it is easy to calculate the rate of division of the cells/tissue in question. This is done by the following formula:

 $Mitotic index = \frac{number of cells undergoing mitosis}{total number of cells}$ 



# **MOLECULAR BIOLOGY**





# 2.7. DNA replication, transcription and translation

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– DNA replication – Transcription: from gene to messenger RNA (mRNA) – Translation: making functional proteins from mRNA sequences

# 2.8. Cell respiration

- Cell respiration: substrates and products - Anaerobic cell respiration - Aerobic cell respiration

# 2.9. Photosynthesis

- Photosynthesis - Light spectrum and chlorophyll

 Production of oxygen by photolysis – The Calvin cycle: using energy to form carbohydrates and other carbon compounds – Rate-limiting factors of photosynthesis



# 2.1 Molecules to metabolism

# 2.1.1 The carbon atom: the core of compounds essential for life

The field of molecular biology aims to explain living processes in terms of the chemical substances involved. The most frequently occurring chemical elements in living things are carbon, hydrogen, oxygen and nitrogen. Carbon in particular is a very important element in the study of living things, as all organic compounds contain this element. It is an atom that can form four covalent bonds and thus allows for the formation of a wide variety of stable compounds to exist. Some of these compounds are essential for life, such as carbohydrates, proteins, lipids and nucleic acids, which perform very important functions in organisms. Further in this chapter we will look at these compounds separately. Below is a diagram of these four types of molecules:



*Skill:* Identify molecular diagrams of these structures as amino acids, sugars (ribose and glucose) and lipids (fatty acids)

*Skill:* Draw molecular diagrams of amino acids (top left), ribose(top right), glucose (bottom left) and a saturated fatty acid (bottom right)



#### 2.1.2 Metabolism

Metabolism is the web of all the enzyme-catalysed reactions in a cell or organism. Metabolic pathways can consist of chains or cycles, and can be anabolic or catabolic.

#### Anabolism

Synthesis of complex molecules from simpler ones, including the formation of macromolecules from monomers by condensation reactions.

Condensation reactions consist of the removal of a water molecule each time a monomer is added to a polymer chain.

E.g. amino acids  $\rightarrow$  polypeptide + water.

#### Catabolism

<u>Example.</u>

The breakdown of complex molecules into simpler ones including the hydrolysis of macromolecules into monomers.

Hydrolysis consists of the addition of water molecules to break down a polymer.

E.g. polypeptide + water  $\rightarrow$  amino acids.

#### Urea: endogenous molecule or artificially produced toxic compound?





In earlier days, organic molecules were believed to be solely synthesized in living organisms. Because urea is an organic compound synthesized in the kidneys as a waste product, it was believed to only be an endogenous molecule. In the early 1800s however, researchers first managed to synthesize artificial urea using silver isocyanate and ammonium chloride. Nowadays it is used as a nitrogen-releasing fertilizer, as well as in the automobile industry and for medical use.



# 2.2 Water

# 2.2.1 Water: molecular and chemical characteristics

Water is an essential molecule for life on Earth. It is a polar molecule that consists of 2 hydrogen atoms and an oxygen atom bound together by covalent bonds. The principle of covalent bonding consists of sharing of electrons between atoms, thus the polarity of the molecule. Water has a slightly positively charged pole where the hydrogen atoms are located and a slightly negatively charged pole where the oxygen atom is located.

Due to the polarity of water molecules, the small negative charge on the oxygen atom has the ability to attract the slightly positively charged hydrogen atoms in nearby hydrogen atoms. This attraction forms hydrogen bonds between molecules and can explain a number of thermal, cohesive, adhesive and solvent properties of this essential molecule.

Figure 2.2: Diagrams showing the molecular structure of water (top) and the formation of hydrogen bonds between two water molecules (bottom)



# 2.2.2 Thermal, cohesive, adhesive, and solvent properties of water

#### **Thermal properties**

High specific heat capacity: large amounts of energy are needed to raise the water's temperature. Hydrogen bonds restrict movement, so more energy is stored by moving molecules of water.

High latent heat of vaporization: hydrogen bonds between water molecules in a liquid form make it very hard for single molecules to escape as vapour. The energy necessary to break these hydrogen bonds and vaporize water is very high compared to other liquids (100 °C). When water vaporizes, a large release of energy occurs, causing a cooling effect on the surface upon which the water used to rest. The concept of sweating as a cooling effect demonstrates this: all the energy used to break hydrogen bonds is released, cooling the skin.



High latent heat of fusion: water at 0 °C must lose a lot of energy before forming ice crystals. Water expands as it freezes and therefore ice can float upon its surface.

#### **Cohesive Properties**

Water molecules can stick to each other due to the formation of hydrogen bonds (due to the polar covalent bonds between the molecule's atoms).

Can explain the formation of water droplets, why some organisms can "walk on water", etc.

#### **Adhesive Properties**

Water can also form hydrogen bonds with other molecules containing oxygen and nitrogen.

#### **Solvent Properties**

Water is an excellent solvent for other polar molecules that attract the charged poles of water molecules (e.g. inorganic molecules with positive or negative charges, polar organic molecules, enzymes, etc.)

This property makes it an ideal transport medium for polar solutes.

# 2.2.3 Hydrophilic vs hydrophobic substances

**Hydrophilic** Substances that are attractive to water and can form intramolecular bonds with it are known as hydrophilic (ionic compounds and polar molecules).

Many hydrophilic compounds can readily dissolve in water.

**Hydrophobic** These molecules are not necessarily repelled by water, they simply are not as attracted to water molecules as much as water molecules attract one another.

These molecules tend to be insoluble in water.



Example.

The hydrophilic and hydrophobic nature of compounds is essential in transport of molecules in blood for example (which has a high water content). Below is the mode of transport of various important molecules based on their solubility in water:

- Glucose and amino acids are polar, so they can be transported and dissolved in blood.
- Cholesterol and fats are non-polar so they are transported in small droplets called lipoproteins, where these non-polar molecules are coated by phospholipids and proteins.
- Oxygen is non-polar, and while some molecules can dissolve in water, they are not sufficient to supply the entire body, therefore, most oxygen is transported in the blood bound to haemoglobin.
- Sodium chloride is an ionic compound. In water, it splits and dissolves as separate Na<sup>+</sup> and Cl<sup>-</sup> ions.

Property	Methane	Water	Fynlanation
Toperty	Mictilalic	water	
Melting point	−182 °C	0℃	Ice melts at a much higher temperature: hydrogen bonds restrict the movement of water molecules and heat is needed to overcome this.
Specific heat capacitty	2.2 J/(g°C)	4.2 J/(g°C)	Water's heat capacity is higher: hydrogen bonds restrict movement so more energy is stored by moving molecules of water than methane.
Latent heat of vaporization	760 J/g	2257 J/g	Water has much higher heat of vaporization: much heat energy is needed to break hydrogen bonds and allow a water molecule to evaporate.
Boiling point	−160 °C	100 °C	Water's boiling point is much higher: heat energy is needed to break hydrogen bonds and allow water to change from a liquid to a gas

#### Comparing thermal properties of water and methane



# 2.3 Carbohydrates and lipids

# 2.3.1 Carbohydrates: from simple to complex compounds

Carbohydrates are complex organic molecules consisting of many monomers called monosaccharides, linked together by condensation reactions.

Two monosaccharides form a disaccharide, more than two monosaccharides form a polysaccharide.

There are several examples of all three classes of carbohydrates:

Monosaccharides: glucose (chemical fuel for cell respiration), fructose (sweet-tasting element in fruit) and galactose.

**Disaccharides:** maltose, lactose (sugar in milk) and sucrose (energy source in plants). **Polysaccharides:** starch, cellulose (main component of plant cell walls) and glycogen

(stores glucose in liver and muscles)

# 2.3.2 Fatty acids: molecular and chemical characteristics

Lipids are hydrophobic compounds that have important functions in:

- Long term energy storage.
- Heat insulation.
- Buoyancy.
- Shock absorption

The main monomer of lipids is the fatty acid long hydrocarbon chains, of which five main kinds exist:

- **Saturated:** all the carbon atoms in the fatty acid chain are connected by single covalent bonds, so the number of hydrogen atoms connected to each carbon cannot be increased.
- **Monounsaturated:** there is one double bond between two carbon atoms in the fatty acid chain.
- **Polyunsaturated:** there is more than one double bond between the carbons in the fatty acid chain.
- **Cis unsaturated:** hydrogen atoms are bonded to carbon on the opposite sides of the double bond.
- Trans unsaturated: hydrogen atoms are bonded to carbon on the same side of the double bond



There are three main classes of lipids: triglycerides, phospholipids (membrane components) and steroids (hormones). We will look at the formation of triglycerides, important in energy storage, by means of a condensation reaction:

3 fatty acids + glycerol  $\rightarrow$  triglyceride + 3H<sub>2</sub>O

Both lipids and carbohydrates are suitable for energy storage, however, several differences in the advantages of each type of compound have been suggested.

#### Carbohydrates

- More easily digested than lipids, good for energy storage that needs to be more rapidly released.
- Soluble in water → easier to transport in blood.

• Can store more energy per gram

Lipids

- than carbohydrates  $\rightarrow$  better for long term energy storage.
- Not soluble in water, also harder to break down and transport around the body (build-up of high energy content fats).

#### Health issues associated to trans- and saturated fatty acids

Trans fats have been banned in several countries in the world, as there has been shown to be a positive correlation between high trans fat content and coronary heart disease.

Saturated fats have also been shown to have a positive correlation (albeit, weaker than trans fats) with the incidence of coronary heart disease.

However, many of the tested populations do not fit these findings, so evidence must be carefully evaluated before banning products and establishing anti trans or saturated fat campaigns.



Example.

#### Determining the body mass index (BMI)

Example.

Because of the natural variation in size between adults, weighing someone does not provide a clear indicator of body mass. Thus, the body mass index was created, and can be calculated using the following formula:

massinka	where:		
$BMI = \frac{mass m kg}{(height in meters)^2}$	Body mass index	Conclusion	
	Below 18.5	Underweight	
	18.5 to 24.9	Normal weight	
	25.0 to 29.9	Overweight	
	30.0 or more	Obese	

A nomogram can also be used to calculate BMI, by drawing a line that connects the height and weight lines, the BMI measure is indicated by the scale in the middle.





# **2.4 Proteins**

# 2.4.1 Amino acids: the building blocks of proteins

Amino acids, containing a carboxyl, an ammine and an R group, are the monomers that when linked together by peptide bonds form complex proteins. Proteins are important organic molecules that carry out major functions in cells and in the extracellular space.

There are 20 different amino acids, which can be linked in any sequence and result in a large range of possible polypeptides.

The specific sequence of each protein is coded for by the genetic material of the organism. As we will see later in the chapter, DNA is transcribed into mRNA and later translated by proteins into polypeptide chains that can combine or form proteins themselves.

Some proteins are made of a single polypeptide while other more complex proteins result from the combination of several polypeptides.

The amino acid sequence also determines the three-dimensional structure of the protein, which will, at the same time, dictate its location and function.

Some proteins are fibrous (long and narrow, usually insoluble), and others are globular (round and 3D, mostly soluble in water).

Polar and non-polar amino acids in these proteins determine the shape and location of the protein, as polar amino acids tend to be exposed to the cytoplasm or extracellular matrix, whereas non-polar amino acids remain in the inner regions of proteins

## 2.4.2 The peptide bond: creating complex amino acid chains that give rise to complex proteins with different functions

Below is a diagram showing the formation of a dipeptide (a 2-amino acid molecule) via a condensation reaction, bound together by a peptide bond, and resulting in the release of a water molecule:





## 2.4.3 Functions of proteins

Below is a table depicting some of the major functions proteins carry out in an organism, as well as specific examples for each function:

Tal	ble	2.1
	~ ~ ~	

Function	Example	Details	Shape
Structural Transport	Collagen Hemoglobin	Strenghen bone, tendon and skin Bind oxygen in the lungs and trans-	Fibrous Globular
Movement	Actin	ports to other tissues Involved in the contraction of mus- cles	Fibrous
Defence	Immunoglobulins	Acts as antibody	Globular

## 2.4.4 Proteomes: the fingerprints of cells

The term proteome refers to the specific proteins expressed by individual cells, tissues or entire organisms. While the genetic make up of an organism is the same in all cells, each tissue or individual cell shows variable gene expression and thus different proteins are created. The proteome of individuals within the same species is quite similar (as the genetic make up is also similar), however, each individual has a unique proteome (like a fingerprint, which can be similar but never identical to other individuals).



# **2.5 Enzymes**

#### 2.5.1 Concepts and definitions

**Enzymes** are long chains of amino acids that give rise to globular proteins. They function as biological catalysts that speed up chemical reactions in cells.

- Substrates are the substances to which enzymes bind and act upon.
- Active site is the region on the enzyme's surface to which substrates bind.
- The activity of enzymes relies on the concepts of molecular motion and collision, in other words, substrates and enzymes must "collide" with one another due to their individual motion (kinetic energy). The more collisions between enzyme and substrate, the faster the reaction occurs.
- A proposed model for enzyme binding is the lock-and-key model: because of the particular shape of an enzyme's active site, only a specific substrate can bind to it (as a key fits a lock). Thus, each enzyme catalyses a specific reaction

# 2.5.2 Influencing enzyme activity: temperature, pH and substrate concentration

#### Temperature



Enzyme activity increases as temperature increases, often doubling with every 10 °C rise. This is because collisions between substrate and active site happen more frequently at higher temperatures due to faster molecular motion.

At high temperatures enzymes are denatured and stop working. This is because heat causes vibrations inside enzymes which break bonds needed to maintain the structure of the enzyme.





Enzyme activity is reduced as pH increases above the optimum because the conformation of the enzyme is altered more and more. Above a certain pH the alkalinity denatures the enzyme and it does not catalyze the reaction at all.

Enzyme activity is reduced as pH increases above the optimum because the conformation of the enzyme is altered more and more. Above a certain pH the alkalinity denatures the enzyme and it does not catalyze the reaction at all.

#### Substrate concentration



At low substrate concentrations, enzyme activity increases steeply as substrate concentration increases. This is because random collisions between substrate and active site happen more frequently with higher substrate concentrations.

At high substrate concentrations most of the active sites are occupied, so raising the substrate concentration has little effect on enzyme activity.

# Example.

#### The use of lactase in the production of lactose-free milk

Many enzymes are very useful in industrial processes (majorly, for instance, in the food industry). Enzymes are often immobilized and employed in large concentrations to catalyse a wide range of biochemical reactions. A common example is the use of enzyme lactase in the production of lactose-free milk.

Lactose is the sugar in milk. Many people are intolerant to this molecule, so often times milk and other milk products are treated with immobilized lactase (an enzyme that breaks down lactose into a maltose and a galactose molecule), and lactose is broken down. The resulting monosaccharides are easier to digest by lactose-intolerant people, and result in a sweeter flavour (less artificial additives needed). The use of the enzyme also speeds up the production of fermented products like yogurt and cheese.


Example.

Immobilized lactase can be used in much larger concentrations and can resist larger changes in pH and temperature compared to endogenous lactase.

The immobilized enzymes can also be reused, and the products are not contaminated with enzymes (easier to introduce and remove from the sites of reaction.

### 2.6 Structure of DNA and RNA

### 2.6.1 Nucleotide structure

Two major types of nucleic acids DNA (deoxyribonucleic acid) and RNA (ribonucleic acid) are essential compounds involved in gene expression in cells. Each nucleotide consists of a 5 carbon sugar linked to a phosphate group at carbon 5, and to one of four nitrogenous bases (adenine, guanine, thymine and cytosine) at carbon 1. The overall nucleotide structure is shown in the diagram below:



Skill: Draw a simple diagram of the structure of single nucleotides (you may use simple circles for the phosphate, pentagons for the sugar [deoxyribose or ribose] and rectangles for the base).



5

### 2.6.2 DNA vs RNA

Both types of nucleic acids share structural similarities, but also significant differences:

RNA	DNA
Contains a 5-carbon sugar	Contains a 5-carbon sugar
Sugar is called ribose	Sugar is called deoxyribose
Single-stranded molecule	Double-stranded molecule
Contains bases adenine (A), uracil (U), cytosine (C), and guanine (G)	Contains bases adenine (A), thymine (T), cytosine (C), and guanine (G)

### 2.6.3 The formation of the DNA double helix

DNA is composed of a double stranded helix of DNA nucleotides. One strand of DNA is held together by covalent bonds that form between the phosphate group of one nucleotide to carbon 3 of the neighbouring nucleotide. This forms a single-stranded backbone. The DNA double strand is then formed by the formation of hydrogen bonds between the nitrogenous bases of two nucleotide strands. Base pairing in DNA is complementary, meaning that one base can only bind to a specific complementary base:

- Adenine (A) binds to thymine  $(T) \rightarrow 2$  hydrogen bonds.
- Cytosine (C) binds to guanine (G)  $\rightarrow$  3 hydrogen bonds.

### **DNA** structure



The two DNA strands are antiparallel, in other words, they run in opposite directions (where one strand has a 5' end, the complementary strand has a 3' end).

The diagram shows the structure of the DNA double helix, showing two joined antiparallel DNA strands bound together by complementary base pairing of adenine with thymine (2 H-bonds); and cytosine and guanine (3 H-bonds).



# 2.7 DNA replication, transcription and translation

### 2.7.1 DNA replication

During the DNA replication process, one double stranded DNA molecule gives rise two daughter DNA molecules. This process is said to be semi-conservative, meaning that each new DNA double helix contains one newly synthesized daughter strand and one strand from the original parent DNA strand, which serves as a template to ensure that both new strands are identical. Essentially, part of the original DNA is conserved at each replication step. Below is a brief description of the process of DNA replication:

- Takes place during the synthesis (S) phase of the cell cycle.
- Helicase unwinds the double helix and separates the two DNA strands by breaking hydrogen bonds.
- The two parent strands that emerge from this process serve as templates for the new daughter strands to be synthesized.
- Enzyme DNA polymerase can then link free nucleotides to the template strands by complementary base pairing.
- Two identical daughter DNA strands are created, resulting in two semi-conservative double stranded DNA helices.

## Taq DNA polymerase: production of multiple copies of DNA by polymerase chain reaction (PCR)

This technique has been one of the greatest biotechnological developments in DNA research. It allows scientists to amplify desired regions of DNA in very little time. PCR consists of the following steps:

- 1. Isolate the desired region of DNA (using restriction enzymes).
- 2. Introduce it in a mixture containing free nucleotides, primers and Taq DNA polymerase.
- 3. The mixture is heated up to 90 °C to separate the DNA strands of the original template.
- 4. Temperature is then reduced to  $55 \,^{\circ}$ C to allow for primer annealing to the now separated strands.
- 5. Taq polymerase (isolated from thermophiles, organisms that can survive at very high temperatures) works optimally at 72 °C, so the mix is heated to this temperature to enhance the formation of new double-stranded copies of the original DNA.
- 6. Process is repeated several times until the DNA is amplified.



# 2.7.2 Transcription: from gene to messenger RNA (mRNA)

Transcription is the synthesis of mRNA copied from the DNA base sequences present in an organism's chromosomes. The sections of DNA that code for polypeptides are called genes, but in order for these polypeptides to be expressed, machinery located outside the nucleus is needed. Thus, a messenger RNA (mRNA) molecule carries the "message" from the DNA to the cytoplasm. Below is the explanation of the process of transcription:

- RNA polymerase unwinds and unzips the area of the DNA to be transcribed (usually containing only one gene).
- RNA polymerase also catalyses the addition of free RNA nucleotides to one of the newly separated DNA strands (this creates a copy of the complementary DNA strand containing the gene of interest).
- In this process, thymine is replaced by nitrogenous base uracil (only present in RNA nucleotides).
- Once the whole gene has been transcribed, the resulting single-stranded mRNA molecule peels off and moves out of the nucleus to be translated into a polypeptide.

### AUCGAACGUUGGGCCCGA

**TIP:** first deduce the complementary base sequence to the given mRNA to get one of the DNA strands, the second DNA strand, as you will realize, is identical to the mRNA sequence, with thymine replacing the uracil bases in the sequence.

# 2.7.3 Translation: making functional proteins from mRNA sequences

Once the DNA "message" has entered the cytoplasm in the form of mRNA, translation takes place and polypeptides are synthesized by ribosomes. When studying this process, researchers found that the genetic code is written in a language of codons, which consist of three consecutive bases (triplet). They found that each codon codes for a specific amino acid (see the table below to see which codons code for what amino acids). Codons are located on the mRNA sequence, while anticodons (complementary to codons) are found on tRNA molecules, another type of RNA that carries the appropriate amino acid to the ribosome where translation occurs. Below is a description of this process:

- The mRNA strand created during the process of transcription binds to a ribosome.
- The ribosome begins to slide over the mRNA until it reaches a starting codon, where a tRNA with a complementary anticodon can bind, coding for a specific amino acid.
- A second tRNA molecule with the correct anticodon binds to the second codon.
- The two amino acids carried by these tRNAs bind to one another by the formation of a peptide bond.



Skill: While you must be able to determine the mRNA sequence that will result from a given DNA sequence, it is essential that you can also go backwards, and deduce the original DNA sequence from a given mRNA sequence. Try it with the following mRNA sequence!

- The ribosome slides over the mRNA molecule, leading to the release of one of the tRNAs (the one that is no longer carrying an amino acid) and the binding of another tRNA to the following codon.
- The amino acid chain keeps growing as this process is repeated until a stop codon is reached, at which point the polypeptide breaks away from the tRNA and can fold and be modified to become a functional protein

Figure 2.5: tRNA & ribosome structure



### Universality of the genetic code: producing human insulin in bacteria

The genetic code has been shown to be universal. That is, in all organisms, the codon code is the same (one codon codes for the same amino acid in any organism). This is very advantageous, as researchers have been able to synthesize important proteins at higher rates by introducing a human DNA sequence for instance in a smaller organism like E. coli, resulting in faster synthesis of the desired protein.

Insulin is a great example of this. Researchers isolated the insulin gene from humans and introduced it into E. coli (a bacterium that rapidly replicates and can yield large amounts of protein in very short time periods). E. coli can then transcribe and translate the insulin gene using its innate machinery. Researchers can then isolate and purify this very important enzyme and use it in for example, the treatment of diabetic patients.

Below is a table showing this universal codon code:

### A C T A C G T A C C T G G G A C T A G A C T

Skill: Use a table of the genetic code to deduce which codon(s) corresponds to which amino acid. For example, try coding the following DNA sequence (only one strand is given) into its transcripted mRNA sequence and this sequence into separate amino acids (remember to first find the start codon, and to correctly identify the stop codon, if present).



### 2.8 Cell respiration

### 2.8.1 Cell respiration: substrates and products

Cell respiration is the controlled release of energy, in the form of ATP, from organic compounds in cells, and follows the equation below:

 $\begin{array}{rcl} \text{Glucose} & + & \text{Oxygen} & \longrightarrow & \text{Carbon Dioxide} & + & \text{Water} & + & \text{ATP} \\ \text{C}_6\text{H}_{12}\text{O}_6 & + & 6\text{O}_2 & \longrightarrow & 6\text{CO}_2 & + & 6\text{H}_2\text{O} & + & 36 \sim 38 \text{ ATP} \end{array}$ 

Cell respiration can follow an aerobic (in the presence of oxygen) and an anaerobic pathway (no oxygen). The latter creates a much smaller yield of ATP.

### 2.8.2 Anaerobic cell respiration

When no oxygen is available to the cells, the following process occurs:

- Glycolysis occurs in the cell's cytoplasm, where a glucose molecule is broken down into two smaller 3-carbon molecules called pyruvate.
- This process leads to a small yield of ATP (2 molecules per reaction) and other products that can later be used in aerobic cell respiration.
- In yeast cells, pyruvate is converted into ethanol and carbon dioxide (there is no further yield of ATP and the products are released as waste). This process is known as fermentation.
- In mammalian cells, pyruvate molecules are converted into lactate molecules (also known as lactic acid), with no further yield of ATP. Lactate accumulates and can lead to changes in pH (lactic acidosis), which can be dangerous in the long term

### 2.8.3 Aerobic cell respiration

When oxygen is present, pyruvate can be further broken down in the cytoplasm and enter the mitochondria in the form of acetyl-CoA (a 2-carbon molecule).

- Acetyl-CoA enters the Krebs cycle, where a series of redox reactions lead to the release of carbon dioxide and the formation of intermediate molecules.
- These molecules are used in the electron transport chain (at the mitochondrial membrane), resulting in a large yield of ATP and the release of water as a by-product



### 2.9 Photosynthesis

### 2.9.1 Photosynthesis

**Photosynthesis** is the process in which plants produce their own organic substances to be used as nutrients.

The process uses energy from the Sun and simple organic compounds (water and carbon dioxide) to create complex carbohydrates to be used as fuel (mainly glucose) and oxygen:

Carbon Dioxide + Water  $\longrightarrow$  Glucose + Oxygen  $6CO_2$  +  $6H_2O$   $\longrightarrow$   $C_6H_{12}O_6$  +  $6O_2$ 

### 2.9.2 Light spectrum and chlorophyll

Sunlight is made up of a range of wavelengths including colors red, green and blue within the visible light spectrum. The smaller the wavelength the more energy is reflected (blue wavelength), and the larger the wavelength the less energy reflected (red). Green color is reflected from medium wavelengths. To absorb and reflect these light waves, specific pigments in plants are needed. The main photosynthetic pigment is chlorophyll; it absorbs red and blue light very well, and reflects mostly green light (thus giving plants their green color). Chlorophyll is located in clusters inside chloroplasts. Below is the absorption spectrum of chlorophyll, showing peaks at the wavelengths easily absorbed by the pigment (blue and red) and a trough on green, the least absorbed wavelength. By looking at the action spectrum (wavelengths of light most used during the photosynthesis reactions) it is clear why chlorophyll is the main pigment in this process: the wavelengths readily absorbed by chlorophyll are majorly used in photosynthesis.



Skill: Draw the absorption spectrum of chlorophyll and the action spectrum for photosynthesis.



### 2.9.3 Production of oxygen by photolysis

Photosynthesis consists of light-dependent and light-independent reactions. The light dependent reactions result in the yield of ATP, oxygen and hydrogen.

Chlorophyll absorbs light energy form the Sun and elicits the process of photolysis, by activating an enzyme that splits water molecules into hydrogen, electrons and oxygen molecules (oxygen is the main by-product of photosynthesis).

Hydrogen and electrons are then involved in the electron transport chain which results in a yield of ATP and intermediate molecules for the light-independent reactions

### 2.9.4 The Calvin cycle: using energy to form carbohydrates and other carbon compounds

The light-independent reactions lead to the formation of complex carbohydrates.

Also known as the Calvin cycle, where ATP and carbon dioxide are used to convert inorganic compounds into organic compounds. This is achieved by carbon fixation, which requires energy from ATP.

### 2.9.5 Rate-limiting factors of photosynthesis

### **Light Intensity**



At low light intensities, rate of photosynthesis is limited.

Photolysis, which requires the absorption of light waves slows down, and thus, so does oxygen and ATP production.

Indirectly limits the light-independent reactions, as ATP is necessary for carbon fixation to occur.

The graph levels off once all the enzymes and reactions are occurring at the highest speed possible.



### **Carbon Dioxide Concentration**



Temperature

Rate-limiting step in the Calvin cycle  $\rightarrow$  carbon cannot be fixed to inorganic compounds and thus glucose production slows down.

Increasing  $CO_2$  concentration increases the rate of photosynthesis, until the photosynthetic enzymes involved in the cycle (e.g. rubisco) reach their saturation point and can no longer increase reaction rates.

Rate of photosynthesis Lemperature

At low temperatures, the enzymes involved in photosynthetic reactions work very slowly.

Rate of reaction increases steadily as temperature increases, until reaching an optimum point when all enzymes are working at a high rate.

When the temperature surpasses this optimal point, enzymes can be denatured, once again decreasing the photosynthetic rate. Skill: Design experiments to investigate the effect of these factors on photosynthetic rates.



### **MOLECULAR BIOLOGY** | Photosynthesis



# **GENETICS**



### 3.1. Genes and chromosomes 48 - Genes - Sickle cell anaemia as an example of a gene mutation - Genome and Human Genome Project - Complexity of organisms and their genomes - Prokaryotic and eukaryotic chromosomes - Karyograms and karyotypes 3.2. Meiosis 53 - Meiosis I - Meiosis II - Meiosis and variation - Failures of meiosis 3.3. Inheritance 59 - Mendel's law of inheritance - Punnett grids - Co-dominant alleles - Sex linkage - Dominant and recessive genetic disorders - Pedigree charts 3.4. Genetic modifications and 64 biotechnology - PCR and gel electrophoresis - Genetic modifications - Benefits and risks of genetic modifications - Clones and cloning



### 3.1 Genes and chromosomes

### **3.1.1 Genes**

Gene a DNA sequence that defines a certain heritable characteristic.

One DNA molecule contains many genes, but not all of these genes are "switched on", meaning expressed.

**Chromosome** a DNA molecule that carries genes.

Within a species, all chromosomes are made of the same DNA molecule, with minor variations of alleles and other mutations.

The expression of genes on this DNA molecule is what differs one chromosome from another.

The number of chromosomes is defined per species. In eukaryotic organisms, chromosomes come in pairs with two chromosomes of the same pair carrying the same genes: a human, for example, has 46 chromosomes, meaning 23 pairs.

Allele a variation of a certain gene, differing from the other allele of the same gene by a few bases.

Alleles are a result of mutations of the gene sequence. Most genes come in two or more allelic forms. Since chromosomes come in pairs, an organism can have two possible alleles of a gene.

If an organism has two of the same alleles, it is called *homozygous* for the trait.

If an organism has two different alleles, it is said to be *hetrozygous* for the trait.

Alleles can be dominant and recessive, where **dominant alleles** are always expressed if present, while **recessive alleles** are only expressed if present homozygously.





Sickle cell anaemia is a heritable disease caused by a mutation of a gene coding for the haemoglobin molecule.



A mutation is a change in the base sequence of a DNA molecule.

In sickle cell anaemia, a base substitution mutation causes a an adenine base in the GAG triplet to be substituted by thymine (GTG).

Recall that a triplet of nucleotides codes for a specific amino acid within a protein.

In this case, GAG codes for glutamic acid which becomes substituted by valine (GTG) in sickle cell anaemia.

The difference in this amino acid changes the shape of the haemoglobin protein, leading to a less functional molecule.

Individuals with sickle cell anaemia have red blood cells with lower oxygen-carrying capacity compared to normal red blood cells.

On the other hand, malaria parasite is less likely to infect the sickle cells compared to the healthy ones, so the people with this condition are resistant to malaria.



### 3.1.3 Genome and Human Genome Project

**Genome** refers to the entire genetic information of an organism.

This means that genome includes more than just all the genes.

Animal cell genome includes chromosomal DNA as well as DNA in the mitochondria, just as a plant genome includes all chromosomes and chloroplast DNA.

Bacterial cell genome includes their chromosomal DNA and their plasmid DNA

Human Genome Project refers to the sequencing of the entire human genome.

HGP characterised all the genes in a human (23,000 of them).

The rest of the DNA sequence is the non-coding region which can control expression of the coding regions.

### 3.1.4 Complexity of organisms and their genomes

The size of the genome of a species does not relate to the complexity of the organism.

Similarly, the number of genes in an organism does not correlate with the complexity of that organism

Organism	Genome size (number of bases)	Number of genes	Number of chromosomes
E. coli	4.6 million	2,300	
Maize (corn)	2.3 billion	32,000	10
Mouse	2.8 billion	23,000	40
Human	3.0 billion	21,000	46





### **3.1.5 Prokaryotic and eukaryotic chromosomes**

### **Cairn's method of measuring DNA length**

Cairn's method involves adding a radioactively labelled nucleotide (thymidine) to cells undergoing replication.

Recall that during replication, each parent strand gains a new daughter strand which assembles from the available nucleotides through homologous base pairing.

In this way, the two new daughter strands incorporate the radioactive nucleotides (thymidine, which also discriminates between DNA and RNA replication) and can be visualized after light exposure.

This method was used to show the nature of replication in bacterial, and subsequently in animal cells.

	DNA	Chromosomes	Plasmid
Prokaryotic	Circular "Naked"	1 circular	Present
Eukaryotic	Linear Associated with proteins	Many linear	Not present

Table 3.2: Comparison of prokaryotic and eukaryotic DNA

### **Prokaryotic DNA**

- Bacteria have one circular DNA molecule that is not associated with proteins.
- Extra genetic information is stored on a plasmid, and can easily be shared between bacteria.
- Antibiotic resistance is often found on plasmid DNA

### **Eukaryotic DNA**

- DNA is associated with proteins called histones.
- Histones are used to wrap DNA around them in order to protect it against damage as well as to control expression of certain genes.
- Recall that eukaryotic chromosomes come in pair.
- A pair of identical chromosomes is called a homologous pair and these chromosomes carry the same genes (with possibly different alleles).
- A complete set of chromosomes (in a human 46) is called a diploid number of chromosomes.
- Sex cells contain half the number of chromosomes (one from each pair), which is said to be haploid, in order to conserve the species' number of chromosomes after fertilization (joining of two sex cells).



### 3.1.6 Karyograms and karyotypes

Karyogram is an image of all the chromosomes of an organism's cell, shown in decreasing size of the homologous pairs.

Karyograms can help determine the sex of the organism as well as the possible chromosomal irregularities that might be disease-causing.

One such irregularity in humans is trisomy 21 (an extra chromosome in the 21<sup>st</sup> pair) that is a cause of the Down syndrome.

23<sup>rd</sup> pair of human chromosomes determine the sex of a baby.

These chromosomes are called sex chromosomes (as opposed to all other ones that are called autologous).

These chromosomes do not have to be identical; a male individual will have one X and one Y chromosome which are of different sizes, while a female individual will have two X chromosomes of the same size (and banding pattern).



ACADEM

Figure 3.1: Karyogram

Karyotype is the characteristic pattern of chromosomes of an organism, referring to their size, shape and the banding pattern.

Kayogram is obtained in two possible ways: amniotic fluid sampling and chorionic villus sampling.

### **Amniotic fluid sampling**

A hypodermic needle is inserted through the abdomen of the mother into the amniotic sack.

The embryo swims in the amniotic fluid which then contains the cells that the embryo has shed off.

By collecting these cells, the karyogram could be obtained.

### **Chorionic villus sampling**

Similarly, the chorionic villi make up the embryonic side of the placenta.

These villi are of the embryonic tissue origin which means that they contain the same cells as the child.

By sampling the chorionic villi (again using a needle), the child's cells can be obtained and karyogram constructed.

### 3.2 Meiosis





### **GENETICS** | Meiosis

The AIM of meiosis is to create cells with half the number of chromosomes, so that during fertilization, each parent could contribute their own set of genes to the offspring and thereby conserve the number of chromosomes of a species and promote variation.

Meiosis consists of two division, where the first one is referred to meiosis I (or reduction division) while the second one, meiosis II can be referred to as mitotic division.

In meiosis I, the number of chromosomes is halved between the two newly formed cells (therefore the name "reduction") while in meiosis II, the number of chromosome between the parent and daughter cells stays the same, but the chromatids separate (therefore the name mitotic).

The stages of each meiotic division have the same name as in the mitotic division, but the events of each stage differ slightly.

Meiosis also involves duplication of DNA, which occurs prior to first meiotic division.



Figure 3.2: Meiosis I

### 3.2.1 Meiosis I

Events in bold differ from mitosis.

### **Prophase I**

- Prior to this phase, the DNA has already duplicated (S phase) and the cell contains double the number of chromosomes (depicted as two chromatids of each chromosome).
- DNA supercoils and chromosomes shorten.
- Nuclear envelope breaks down to allow.
- Centrioles move to the poles.



### **Metaphase I**

- Homologous chromosomes pair up at the equator (this means that the two chromosomes of each pair, line up on top of each other, rather than next to each other like in mitosis).
- Each pole's spindle microtubule attaches to one chromosome from each homologous pair (recall that in mitosis, one chromosome would have one of each pole's spindle microtubules)

### **Anaphase I**

- Each spindle microtubule pulls one whole chromosome of the homologous pair towards it's pole, causing a division of the chromosome pairs across the cell (recall that in mitosis, at this step, only the chromatids would separate so that a full set of chromosomes would be present at each pole).
- The movement of the **chromosomes** is achieved through shortening of the spindle microtubules.

### **Telophase I**

- Nuclear envelope forms around each set of chromosomes.
- The cell divides into two cells with **haploid number of nuclei** (only one chromosome from each pair).
- Chromosomes partly uncoil.
- The cell will proceed with meiosis II

### 3.2.2 Meiosis II

Figure 3.3: Meiosis II



ACADEMY

Note that the events of this phase are identical to mitosis, but the starting number of chromosomes is halved!

Before meiosis II, there is no duplication of DNA, so the cell proceeds straight from the telophase into the new division.

Recall that each cell now possesses one chromosome of each pair, but with two chromatids (meaning that each chromosome is made up of two identical DNA molecules!)

### **Prophase II**

- Chromosomes supercoil again and become shorter.
- Centrioles again move to the poles of the cell.
- Nuclear envelopes break down.

### **Metaphase II**

- Chromosomes line up at the metaphase plate, one next to each other across the equator.
- Spindle microtubules (one from each pole) attach to the centres of the chromosomes (centromeres).

### **Anaphase II**

- Spindle microtubules pull the chromatids apart, so that one chromatid of each chromosomes travels to the opposite pole.
- Therefore, each pole of the cell will receive one DNA copy of each chromosome.

### **Telophase II**

- At this last stage, each pole of the cell contains half the number of chromosomes compared to beginning of meiosis I, but the same number of chromosomes (just half the chromatids) compared to meiosis II.
- The nuclear envelope forms and the cell divides into two cells.
- Recall that in the first meiotic division, two cells were formed, meaning that now, each of those two cells divided into two, yielding a total of 4 cells, each with half the number of chromosomes



### 3.2.3 Meiosis and variation

When we talk about variation and meiosis, it is important to understand the following terms:

**Crossing over** process through which the non-sister chromatids within a homologous pair, exchange genetic material during prophase I.

Through exchange of genetic material between non-sister chromosomes of a homologous pair, gametes end up with chromosomes with different allele combinations from their somatic cells.



Recall that each chromosome possesses different genes from other chromosomes and possibly different alleles from their homologous pair.

Depending on which chromosome orients towards which pole in metaphase 1, each cell will end up with a different set of chromosomal alleles.

If these definitions are clear, then it is obvious how within one individual, there is an infinite variation between the gametes. Now, adding the process of fertilization, where two random gametes from two different individuals of a species fuse, there is truly an infinite number of possible combinations and therefore infinite genetic variation.



### 3.2.4 Failures of meiosis

Recall that failures of checkpoint mechanisms during mitosis often result in tumour formation. The failures of meiosis, on the other hand, are usually lethal for the embryo, or result in genetic disorders.

### Down Syndrome

Example.

Down syndrome is a chromosomal abnormality in which the individual possesses 1 extra chromosome in their 21<sup>st</sup> chromosomal pair.

This is a result of improper separation of homologous chromosomes during anaphase I, or improper disjunction of sister chromatids during anaphase II.

The cell that ends up with an extra chromosome in the 21<sup>st</sup> pair will be able to continue its life cycle through fertilization.

The cell that ends up with one less chromosome will not be able to continue its life cycle and will not survive past fertilization.

Disjunctions are believed to be common, but most of them are lethal to the embryo and present themselves as spontaneous abortions.



### 3.3 Inheritance



### 3.3.1 Mendel's law of inheritance

Mendel discovered some basic laws of inheritance by cross-fertilizing pea plants with different traits (flower colour, pea shape). He observed "hidden" traits that tend to surface after several generations. These were in fact what we now call recessive alleles.

Since gametes possess only one set of chromosomes (haploid) compared to somatic cells, they also possess only one allele that they can pass onto the offspring. The other parent will give the other allele of the gene.

Since alleles are variations of traits (e.g. a gene codes for hair colour in general, but the alleles code for the specific hair colours), the combination of alleles will determine the final trait of the individual.

A punnett grid is a useful tool to predict all the possible offspring combinations of a particular trait.



### 3.3.2 Punnett grids

Represent the maternal alleles on one side, and paternal alleles on the other side of the grid.					
In the first step, based on the parent's full set of			A	В	
chromosomes, one can determine what possible alleles	-			_	
the parents' gametes can have.	nal es	С	AC	BC	
In the second step, by combining all the possible parents'	lel				
alleles in the grid, one can determine the possible	pat al	D	AD	BD	
offspring combinations.					

### Rules of punnet grids

Parental generation is called P1, first offspring generation F0 and all other offspring generations are numbered F1, F2 etc.

Dominant alleles determine the letter used to describe the trait (if brown eyes are dominant to green eyes, than the trait will be defined by letter B and not G).

Dominant alleles are written in the capital letter, and recessive alleles in the lower case letter (therefore, brown eyes will be B, and green eyes will be b, not g!).

### Pea flower colour (purple allele dominates the white allele)

	Parent 1	Parent 2
Genotype	PP (homozygous dominant)	pp (homozygous recessive)
Possible gametes	P and P	p and p
Phenotype	Purple flowers	White flowers

The punnet grid is constructed by placing the alleles of one parent on the vertical side of a  $2 \times 2$  square, and the other parent on the horizontal side.

		Pare	nt 1		
		Р	Р	P1:	PP pp
~ <sup>_</sup>				F0 Genotype	Heterozygous (dominant) Pp
nt	р	Рр	Рр	F0 Phenotype	Purple (since one dominant allele is always
Pare	р	Рр	Рр		present)

Let's see now what happens if you cross-pollinate the offspring.

		Pare	nt 1		
		Р	b	P1:	Pp Pp
~ <sup>–</sup>			1	F1 Genotype	25% PP, 25% pp, 50% Pp
nt 2	Р	PP	Рр	71	(25% HomD, 25% HomR, 50% HetD)
Pare	р	Рр	рр	F1 Phenotype	75% purple, 25% white



Example.

### 3.3.3 Co-dominant alleles

Recall that co-dominant alleles are those where none of the alleles over-rules the others. All the present alleles are expressed.

Each individual has one of the four possible blood groups, namely A, B, AB or O. These are in fact the surface molecules carried on the red blood cells that help the body distinguish between self and non-self. If an individual has surface molecules A, then the body will be trained to recognize all the B molecules, as well as the lack of molecules (O) as foreign. This is why it is so dangerous to receive blood from a mismatched donor.

Alleles that determine the blood groups are either allele for A, for B, or for O. Alleles A and B are co-dominant, and allele for O is recessive to both A and B. Therefore, if an individual has both the allele for A and B, her blood group will be AB, but if she has allele A and allele O, her blood group will be A.

### **Rules of punnet grids**

- For co-dominant alleles, the letter used to represent all alleles is capital I.
- For the specific alleles, for example in the ABO blood group example, blood group A is labelled as capital I with a superscript A, so I<sup>A</sup> and blood group B as I<sup>B</sup>.
- If there is another allele that is recessive to both the co-dominant alleles, that one is labelled as a lower case letter i with no superscript.

### ABO blood groups

Example.

	Parent 1	Parent 2
Phenotype	Blood group A	Blood group B
Genotype	I <sup>A</sup> i (heterozygous A)	I <sup>B</sup> i (heterozygous B)
Possible gametes	I <sup>A</sup> and i	I <sup>B</sup> and i

		Pare	nt 1			
		IA	i	P1:	I <sup>A</sup> i I <sup>B</sup> i	
nt 2	$\mathbf{I}^{\mathrm{B}}$	I <sup>A</sup> I <sup>B</sup>	iI <sup>B</sup>	F1 Genotype	25% I <sup>A</sup> i, 25% I <sup>B</sup> i, 25% I	<sup>A</sup> I <sup>B</sup> , 25% ii
Pare	i	I <sup>A</sup> i	ii	1 <sup>-1</sup> Thenotype	25 % A, 25 % D, 25 % AD	and 23 /0 U

### Another example

	Parent 1	Parent 2
Phenotype	Blood group AB	Blood group 0
Genotype	I <sup>A</sup> I <sup>B</sup> (co-dominant)	ii (homozygous recessive)
Possible gametes	I <sup>A</sup> and I <sup>B</sup>	i and i





### 3.3.4 Sex linkage

Recall that the chromosomes can be divided into somatic and sex chromosomes. The sex chromosomes are the ones that determine the sex of an organism.

Sex chromosomes are labelled as X and Y. Y chromosome is much smaller: for that reason, X chromosome carries more genes that the Y chromosome does.

The presence of Y chromosome determines that gender of the child will be male: females normally carry two X chromosomes, while males carry one X and one Y chromosome.

This means that the female will pass on only the X chromosomes to her offspring, while the male can pass on either an X or a Y chromosome.

In order for a female offspring to be born, gametes carrying an X in both male and female cell must meet, and for a male to be born, a female gamete with X and a male gamete with Y have to fertilize.

Sex linkage refers to the inheritance of genes that are located on the sex chromosomes.

Since the X chromosome carries more genes than the Y chromosomes, males will often lack one copy (and therefore one allele) of the sex linked genes. For this reason, many of the sex linked diseases affect males to a higher degree than females.

### **Rules of punnet grids**

- For sex linked genes, the letters assigned to the traits are either X or a Y, depending on the gender of the individual.
- The trait is labelled as a superscript on the X or Y.
- Dominant allele is written in the capital letter, while the recessive is written in the lower case letter.



Red-green colour-blindness is a sex linked disorder carried on the X chromosome where the affected individuals cannot distinguish between red and green colours. It is a recessive disorder, meaning that only individuals with no dominant (healthy) alleles are affected by it. However, since it is a sex linked trait, carried on the X chromosome, males already have a disadvantage since they have only one chromosome that can either carry the healthy or the affected gene.

Females carry two X chromosomes, and therefore two alleles for colour vision. Males carry one X chromosome with the gene for colour vision, and a Y that doesn't contain the gene at all. If a female has one healthy and one affected gene, she will be called a carrier but she will be healthy. She might pass on her affected X chromosome to her son, who will not inherit another X chromosome, but a Y chromosome from his father and therefore be affected by the disease. Note that females can be affected as well, but in that case they would have to have a carrier mother and an affected father (and still there is only 50% chance that they will have the disease).

Let's look at what happens in the punnet grid.

Parent	Mother	Father
Phenotype Genotype	Healthy (carrier) X <sup>H</sup> X <sup>h</sup>	Healthy X <sup>H</sup> Y
Possible gametes	$\mathrm{X}^{\mathrm{H}}$ and $\mathrm{X}^{\mathrm{h}}$	$X^{\rm H}$ and $Y$

	Parent 1		P1:	I <sup>A</sup> i I <sup>B</sup> i	
		$  X^H   X^h$		F1 Genotype	$25\% X^{H}X^{H}, 25\% X^{H}X^{h}, 25\% X^{H}X^{h}, 25\% X^{H}X^{h}, 25\% X^{H}X^{h}X^{h}, 25\% X^{H}X^{h}X^{h}X^{h}X^{h}X^{h}X^{h}X^{h}X^{h$
$\frac{1}{2}$	X <sup>H</sup>	X <sup>H</sup> X <sup>H</sup>	X <sup>h</sup> X <sup>H</sup>	F1 Phenotype	75% healthy child, 25% affected
Pare	Y	X <sup>H</sup> Y	X <sup>h</sup> Y		affected, and 0% chance that the
					daugher will be affected)

### 3.3.5 Dominant and recessive genetic disorders

So far, the disorders we have shown have all been recessive. This means that only the individuals with two affected alleles get the disorder. If the disease is of dominant inheritance, one affected allele is enough for the individual to carry the disease. This will result in a much higher percentage of affected individuals in a family tree (compared to the recessive disorders).



			Par	ent 1		Parent 2	
Phenotype Genotype Possible gametes			Hea Hh es hai	Healthy Hh (homozygous recessive) h and h		Huntington's Hh (heterozygous dominant) H and h	
		Pare	ent 1				
Parent 2		h	h	P1:	I <sup>A</sup> i	$I^{B}i$	
	Н	hH	hH	FI Genotype F1 Phenotype	50% Hh and 50% hh 50% affected offspring,		
	h	hh	hh	<i>,</i> 1	50% healthy offspring		

### Huntington's disease

Example.

### **3.3.6 Pedigree charts**

Pedigree charts are a way to represent the inheritance of certain traits in a form of a family tree, where the oldest individuals are set at the top, and their offspring follow downwards.

There are some rules you should keep in mind:

- The female is always labelled as a circle  $\bigcirc$  and the male as a square  $\square$ .
- Most pedigree charts show affected individuals as coloured figures and healthy and carrier individuals as transparent figures .
- Such a chart helps determine the possible genotypes of individuals and chances for affected offspring in the future.

Note that in the exam, you could be asked to also determine whether the disease is sex linked, recessive or dominant.

Here are again some tips that might help you out with that

- In the charts where mostly males are affected usually represent a sex-linked trait (this means that more than 90% of the affected individuals are males).
- Two healthy individuals cannot have a child with a dominant disorder.
- In recessive disorders, two healthy parents can have an affected child, but two affected parents cannot have a healthy offspring.

Figure 3.5: Recessive vs dominant chart



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### **3.4 Genetic modifications and biotechnology**

### 3.4.1 PCR and gel electrophoresis

A single cell contains a miniature amount of DNA. A DNA sample, obtained from a piece of tissue or bodily fluid is often not enough on its own to give conclusive results. For that reason, the DNA needs to be multiplied to make sure there is enough for proper analysis:

### Polymerase chain reaction (PCR)

PCR is a method through which a molecule of DNA is copied many times, yielding a high number of identical DNA molecules. The system is based on the cellular replication process but employs the use of the polymerase enzyme from Thermus aquaticus, which works best at high temperatures. At high temperatures, the two strands of DNA molecules naturally separate, and the polymerase enzyme is able to replicate both chains. When this process finishes, the temperature is lowered, so that the chains join again. This is repeated through several cycles, until the DNA is sufficiently quantified. You can imagine this goes quite fast, since from one DNA molecule, you get two, which can then be amplified to another four, then 16 an so on.

### **Gel electrophoresis**

This method was developed to visualize the DNA fragments, or proteins isolated from cells. The principle of this method is to separate DNA fragments or proteins from a mixture, based on their size and charge. The electrophoresis chamber consists of an agarose gel submerged into a liquid, and two electrodes (a positive and negative) at each side. The voltage across the chamber makes the molecules of different sizes and charges move across the gel at different speed. Large and less charged molecules stick behind, while the small and highly charged molecules travel faster.

The result is a pattern of bands visualized under UV light which can be compared between cells, or individuals (in case of paternity tests).

Gel electrophoresis can be used to determine the paternity of a child by comparing DNA fragments of the mother, the child and the suspected fathers.

Since the child's DNA is a combination of his father's and mother's DNA, the child's patter will contain some of his mother's and some of his father's bands



### **GENETICS** | Genetic modifications and biotechnology

Example.	Paternity determination									
	Mother	Child	Father 1	Father 2	Father 3					
		_			_					



### 3.4.2 Genetic modifications

Recall that the genetic code is universal, meaning that all the organisms translate the same triplets of bases into the same amino acids. With that in mind, you can see why a gene in one organism's coding for a specific protein, could be transferred to another organism's where it would again code for the same protein.

### E.coli and the production of insulin

Bacteria can be genetically modified to produce a human protein of interest. An example of this is the production of insulin through E.coli. This is achieved by extracting the messenger RNA from human cells producing insulin (pancreatic cells) and converting that mRNA back into DNA using reverse transcriptase enzyme.

This piece of DNA could now be ligated into a plasmid using restriction enzymes, enzymes that are able to cut DNA at a specific point with a specific patter, leaving sticky ends through which the gene of interest (from previous step) could be ligated into the plasmid (which has also been cut with the same restriction enzymes).

Once the plasmid with the gene of interest is made, it can incubated with the bacteria which will then take it up and start transcribing and translating the newly acquired gene. These bacteria are now called recombinant bacteria.

The result will be the production of the protein, which can then be collected as a part of the bacterial secretions.





### 3.4.3 Benefits and risks of genetic modifications

### **Benefits**

- With less pest damage, there are higher crop yields, so less food shortages.
- With better yield, less land is needed for the same amount of crop production, and the unused land can be conserved for wildlife.
- There is no need to use pesticides that damage other organisms living in the vicinity.

### Risks

- Long term effects of genetically modified foods have not yet been determined.
- The pollen of the modified crops might be blown away to the wildlife where it might kill the organisms that do not normally infect the plant.
- The genetically modified plants have an evolutionary advantage to the non-modified plants, so the random cross-pollination might create an imbalance in the ecosystem.

# Example.

### Bt maize

Bt maize can be genetically modified to express a bacterial gene for Bt toxin that is toxic to the pests that usually attack it. In that way, the crops are "resistant" to the pest infestations.

### 3.4.4 Clones and cloning

**Clone** an organism that is genetically identical to its parent organism.

Cloning is a technique of producing genetically identical cells, tissue or organisms. This is usually done to obtain a higher number of cells or individuals with a desirable set of characteristics. Cloning of plants can be relatively easy by taking a piece of root or stem that contains plant stem cells that will grow into a new, genetically identical plant. Animal cloning is more difficult since animals cannot develop from a group of stem cells found in the body.



### Dolly the sheep

Dolly the sheep was the first cloned animal. 3 sheeps were used to give rise to Dolly, namely one that donated the egg cell (without the nucleus), one sheep that donated its genetic material (nucleus of a somatic cell) and one sheep into which the embryo was implanted. Therefore, Dolly was genetically identical only to the sheep donating the nucleus.

### Process

An egg cell was obtained from a sheep, and the nucleus of that cell was removed. The egg cell contains only half the genetic information, so its nucleus cannot be used in cloning. However, the egg cell contains the vital enzymes and organelles for development.

Udder cells (somatic cells) from another sheep were taken and grown in deprived environment which cause them to switch off all their genes. A nucleus of such a cell was isolated (it contained all the chromosomes), and then fused with the egg cell without the nucleus.

The fused cells were inserted into the third sheep (the surrogate) and they developed as a newly formed embryo. The sheep that was born was named Dolly and was genetically identical to the mother providing the nucleus of its cell.

Cell taken from udder of donor adult and cultured in laboratory for six days



Unfertilized egg taken from another sheep. Nucleus removed from the cell

Egg without a nucleus fused with donor cell using a pulse of electricity



Surrogate mother gives birth to lamb. Dolly is genetically identical with the sheep that donated the udder cell (the donor)

Embryo resulting from fusion of udder cell and egg transfered to the uterus of a third sheep which acts as surrogate mother



Example

**GENETICS** | Genetic modifications and biotechnology



# **ECOLOGY**



# 4.1. Species, communities and ecosystems - Autotrophs vs. heterotrophs 4.4. Species and definitions - Autotrophs vs. heterotrophs 4.4. Heterotrophs: consumers vs. detritivores vs. saprotrophs 4. Nutrient cycling 4.2. Energy flow 4. Sunlight: main source of energy in ecosystems - Food chains and energy pyramids: analyzing energy flow in ecosystems - Energy loss: limiting food chain length 4.3. Carbon cycling 4.4. Carbon cycling A. Carbon cycling A. Carbon of organic compounds

### 4.4. Climate change

 The greenhouse effects and greenhouse gases – The enhanced greenhouse effect and its effect on global temperatures – Changes in atmospheric CO<sub>2</sub> concentrations and average global temperature 76

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### 4.1 Species, communities and ecosystems

### 4.1.1 Key terms and definitions

Species a group of organisms that can interbreed and produce fertile offspring. Its members can be reproductively isolated (live in a different habitat) but as long as they hold the ability to potentially interbreed with other members from different habitats, they are for all effective purposes part of the same species.
Habitat the environment in which a species normally lives, or the location of a living organism.
Population a group of organisms of the same species who live in the same area at the same time.
Community a group of populations living and interacting with each other in the same geographical area.
Ecosystem community and its abiotic environment.
Ecology the study of the relationship between living organisms and between organisms and their environment.

### 4.1.2 Autotrophs vs. heterotrophs

One way organisms can be classified into different groups is based on their method of nutrition, in other words, by the ways in which they obtain organic compounds necessary to provide essential nutrients to each organism. In this way, organisms can be classified as autotrophs and heterotrophs.

Autotrophs can synthesize their own organic compounds from simple organic compounds found in the abiotic environment, to do this, they absorb carbon dioxide, water and other inorganic nutrients from the environment to synthesize organic compounds (e.g. plants, algae, cyanobacteria (all of these are photosynthetic organisms)).

Heterotrophs obtain organic compounds from other living organisms (e.g. humans, insects, fish, etc.).

Some species can obtain nutrients through both methods (e.g. some bacteria) but this is a rare occurrence.


### 4.1.3 Heterotrophs: consumers vs. detritivores vs. saprotrophs

Within the group of heterotrophs, those organisms that obtain organic compounds by ingestion of other living organisms, there are also several subcategories:

**Consumers:** organisms that obtain organic matter from other living or recently killed organisms

- Primary consumer: eats autotrophic organisms  $\rightarrow$  herbivore
- Secondary consumer: eats herbivores  $\rightarrow$  carnivore
- Tertiary consumer: eats secondary consumer → top carnivore (top carnivore can also be a quaternary consumer, depending on the size of the food chain)
- **Detritivores:** decomposers that internally digest dead organic matter like leaves and carcasses (e.g. earthworms and woodlice)
- Saprotrophs: decomposers that live in or on dead organic matter and externally digest it, by secreting digestive enzymes and absorbing the products of digestion (e.g. fungus and bacteria)

### 4.1.4 Nutrient cycling

Since autotrophs sustain themselves by absorbing nutrients and other inorganic compounds from the abiotic environment, there has to be a way in which this inorganic compound supply is maintained. This is achieved through a process known as nutrient cycling. When an organism dies, decomposers are in charge of breaking it down and absorbing as many nutrients and as much energy as possible via digestion. The remains of the organism are then mostly inorganic compounds that return to the abiotic environment after the process of decomposition. Gas balance (oxygen-carbon dioxide) is usually replenished by the processes of cell respiration and photosynthesis in plants and consumers. This returns balance to the ecosystem, once again providing available inorganic matter for autotrophs to make use of. This results in ecosystems having the potential to be sustainable for long periods of time, as long as a source of energy and the necessary fuel sources are available.



#### Mesocosms: proving that ecosystems are self-sustainable

The picture below shows one possible design of a mesocosm; an experimental setup that tries to emulate a real-life ecosystem in an isolated space to determine if the ecosystem is indeed self-sustainable:

Figure 4.1: Mesocosms proving that ecosystems are self sustainable



The setup contains the main elements found in ecosystems: a variety of autotrophs and different types of heterotrophs (including decomposers that enable nutrient cycling), an abiotic environment (mud as a source of inorganic compounds and nutrients, water, air containing oxygen and carbon dioxide. With a source of energy (light), this sealed mini-ecosystem should be able to self-maintain for a long time.

## 4.2 Energy flow

# 4.2.1 Sunlight: main source of energy in ecosystems

In any given food chain, the first and most abundant level of organisms comprises autotrophs (e.g. plants and algae), also known as producers. Since autotrophs are mainly photosynthetic organisms, they require sunlight as a main source of energy in order to convert it into organic compounds and obtain the necessary nutrients for survival. Without this essential energy source, producers would not survive and primary consumers would not be able to feed, severely disrupting the food chain.



# 4.2.2 Food chains and energy pyramids: analyzing energy flow in ecosystems

A food chain is a sequence showing the feeding relationships and energy flow between species sharing a habitat. The arrow shows the direction of the energy flow (energy is taken up upon ingestion by the next organism in the food chain).



A series of interconnected food chains creates a food web, these are much more complicated to draw and interpret (some organisms can be different types of consumers for example). An organism's position in a food chain or web is also known as tropic level, and it is an easy way to classify organisms, the names of each trophic level are shown in grey above the food chain boxes. Energy in the form of carbon compounds flows from one organism to the next when the organism in the higher trophic level feeds on another. However, this energy flow is not 100% effective. We will see why in the section below.

# 4.2.3 Energy loss: limiting food chain length

Energy enters the ecosystem as sunlight, and is absorbed with about 20% efficacy by autotrophic producers and transformed into chemical energy

Chemical energy obtained from photosynthesis is transferred from one trophic level to the next by means of feeding, when carbohydrates, lipids, proteins and nucleic acids are digested

At each step, only about 10% of the energy is successfully transferred to the feeding organism, energy is lost in several ways:

- Energy released from carbon compounds is used in cell respiration and lost as heat
- When feeding, not the entire organism is eaten (bone, cartilage, etc. is often left to decompose)
- Loss of carbon dioxide, water and other waste products throughout the organism's life
- Some organisms die and decay before being eaten



#### ECOLOGY | Carbon cycling

• Warm-blooded and moving organisms lose more energy as heat

While nutrients can be recycled by decomposers, once energy leaves a food chain it cannot be taken up again

Energy losses of this kind restrict length of food chains and the biomass of the higher trophic levels

A pyramid of energy (example shown below) shows the flow of energy from one trophic level to another in a community

- The first step is always the largest (producers)
- At each increasing trophic level, the bar becomes 10 times smaller (scaling is important here), as only 10% of energy is transferred
- Energy flow is expressed in units: kJ/m<sup>2</sup>/year (kilojoules per square meter per year)



#### Figure 4.3: Food chains

## 4.3 Carbon cycling

### 4.3.1 Carbon: how does it enter ecosystems?

Carbon often enters an ecosystem in the form of carbon dioxide, an inorganic molecule that is diffuses into autotrophic producers.

Autotrophs then have the ability to convert carbon dioxide into complex carbohydrates (organic molecules) and other carbon-containing molecules (e.g. proteins and lipids) through the process of photosynthesis.



#### ECOLOGY | Carbon cycling

Carbon dioxide is present in air; so terrestrial autotrophs can directly absorb it through specialized porous cuticles.

In aquatic ecosystems however, carbon can be found as dissolved  $CO_2$  or in the form of  $HCO_3$  (hydrogen carbonate) ions, which result from the chemical reaction when water and carbon dioxide combine to form carbonic acid ( $H_2CO_3$ ).

These molecules can then be used in photosynthesis to produce organic compounds. These organic compounds are then used in the process of cell respiration, leading to the regeneration of carbon dioxide, which is released into the water or the atmosphere as a by-product.

### 4.3.2 The carbon cycle

As stated earlier, carbon dioxide enters ecosystems by diffusing into carbon dioxide, and is transformed into organic compounds through photosynthesis. Some carbon dioxide returns to the atmosphere as a by-product of cell respiration that takes place in producers. Primary consumers feed from producers, taking up some carbon-containing compounds (and secondary consumers feed on primary, etc.).

Cell respiration leads to further generation of carbon dioxide that is released into the atmosphere.

When consumers die, carbon flux reaches decomposers (which also release carbon dioxide as a product of cell respiration)

- A particular group of prokaryotes called methanogenic bacteria can break down organic compounds in anaerobic conditions, leading to the release of methane into the atmosphere, these bacteria act in places where saprotrophs and detritivores can only partially decompose organic matter
- Methane can accumulate in the ground or diffuse into the atmosphere, where it can be oxidized to carbon dioxide

Partial decomposition and fossilization lead to the formation of fossil fuels (more details in the next section)

Combustion of fossil fuels in industrial factories produces carbon dioxide that is released into the atmosphere in great quantities



#### ECOLOGY | Carbon cycling



Skill: Construct a diagram of the carbon cycle.

# 4.3.3 Decomposition of organic compounds

#### Formation of peat and coal

In acidic and anaerobic conditions (e.g. swamps and bogs), saprotrophs cannot fully break down dead organic matter.

Partially decomposed organic matter accumulates to form thick deposits called peat.

Peat can then be crushed and converted into coal, which is a source of fuel that releases carbon dioxide into the atmosphere.

#### Formation of oil and gas

Silt and the remains of dead organic organisms can sometimes accumulate in shallow seas.

Due to the anaerobic conditions, these organisms are only partially decomposed.

This silt accumulates and is converted to shale with certain compounds becoming oil or gas and remaining trapped in the pores of rocks.



# 4.4 Climate change

## 4.4.1 The greenhouse effects and greenhouse gases

The greenhouse effect, despite the negative connotations associated to the concept, is a naturally occurring phenomenon that has enabled life on Earth for millennia.

Sunlight enters the atmosphere in the form of waves. Short-wavelength radiation is partly absorbed by the ozone layer (mostly ultraviolet  $\rightarrow$  about 25%). The remaining 75% of larger wavelengths radiation reaches the Earth's surface, where it is absorbed and produces heat.

The Earth's surface re-emits radiation (like a reflection) at much longer wavelengths (infrared  $\rightarrow$  heat).

A high percentage (75–80%) of this radiation is absorbed by greenhouse gases in the atmosphere.

These gases re-emit radiation, and some of it reaches the surface of the Earth again, causing warming (atmospheric temperature is significantly warmer that outer space.

Greenhouse gases are special in that they have the ability to absorb this long-wavelength radiation that is emitted from the Earth's surface.

The main greenhouse gases are carbon dioxide and water vapor both found in low concentrations in the atmosphere.

Oxides of nitrogen and methane are also greenhouse gases present in the atmosphere.

It is important to realize that ozone is not a greenhouse gas, so ozone depletion does not increase the greenhouse effect.

## 4.4.2 The enhanced greenhouse effect and its effect on global temperatures

Currently, increased levels of greenhouse gases in the atmosphere caused by industrialization cause the atmosphere to retain more and more heat. This is known as the enhanced greenhouse effect, a phenomenon that has affected average global temperatures and climate patterns on Earth.

Human activity has considerably increased the production of greenhouse gases (predominantly carbon dioxide) in the last 200 years, starting after the onset of the industrial revolution, where the combustion of fossil fuels became a major source of fuels and energy for human development.



#### ECOLOGY | Climate change

Some human activity that has led to the increase in greenhouse gas production:

- Burning of fossil fuels
- Use of ammonia-based fertilizers
- Industrial processes (e.g. production of nitric acid)
- Waste disposal in landfills
- Production and distribution of natural gas

These processes have led to a considerable increase in global temperatures in the last two centuries, leading to concerns about increasing sea level, destruction of arctic habitats, coral reefs, etc.

# 4.4.3 Changes in atmospheric CO<sub>2</sub> concentrations and average global temperature

Over the last 150 years, carbon dioxide emissions have increased significantly, mostly as a result of fossil fuel combustion in industry. While there is a clear correlation between an increase in carbon dioxide emissions and an increase in global temperatures, other variable factors influence this process (e.g. atmospheric concentration of other greenhouse gases).



# **EVOLUTION AND BIODIVERSITY**



<ul> <li>5.1. Evidence of evolution and natural selection</li> <li>- Evidence of evolution - Natural selection</li> </ul>			
<b>5.2. Classification of biodiversity</b> <ul> <li>Binomial system and dichotomous key – Domains and phylogeny – Classification of plants – Classification of animals</li> <li>Classifications of vertebrates</li> </ul>	86		
<ul> <li><b>5.3.</b> Cladistics         <ul> <li>Human classification through cladistics – Reclassification of figworts – Cladogram analysis</li> </ul> </li> </ul>	91		



# 5.1 Evidence of evolution and natural selection

**Evolution** is the accumulation of changes in heritable characteristics of a species.

This means that over times, certain characteristics of a species change, and if these characteristics are inheritable, then this change is transferred to all the subsequent generations.



If the members of one species become separated for a long period of time, their characteristics (due to adaptations to the new environment) will start changing as well.

Sometimes it happens that these two populations change so much that, even if they got reunited, they would not be able to interbreed anymore.

## 5.1.1 Evidence of evolution

#### **Pentadactyl limb**

Mammals, birds, reptiles and amphibians all have limbs which they use to different purposes.

Some limbs are used for running, walking, swimming or jumping, but eventually, all possess the same pentadactyl structure (5 digits).

The explanation for this is that all these animals have the same ancestor that had a pentadactyl limb, but as the organisms adapted to different environments (water, forest, desert, etc.), the exact structure started differing.

As the limb structure changed as a response to different environments, this type of evolution is called adaptive radiation.

#### **Selective breeding**

Domesticated animals are an example of a "fast forwarded" evolution.



Wild animals with favourable characteristics for humans were bred with other such animals of the same species to get offspring with similar characteristics.

This process is called selective breeding.

An example of this would be a dog, which is a domesticated wolf.

Wolves that were less aggressive were domesticated and bred until more such offspring were produced.

Then, for example, only the smaller individuals were bred which resulted in a smaller dog species (etc.)

#### **Fossil records**

Fossils are remaining of organisms found in stones that can help us determine their age and compare them to the currently living species.

It is possible to determine the age of stones where fossils were found and this has shown that bacteria are found in the oldest fossils, followed by algae, fungi and more complex organisms later.

Acanthostega is a fossil of a vertebrae that doesn't match any of the current living species, but shows similarities with them.

It has both 4 limbs that matches amphibians, but also a fish-like tail and gills which suggests it was probably a transition species between the fish and amphibians.

#### Melanism

Melanism refers to the phenomenon where a lightly coloured species has a darker variant.

Lightly coloured moths are well adapted to avoid predators by blending with the lightly coloured tree branches.

The melanistic form cannot blend in with the trees so are frequently eaten.

During industrial revolution, the smog caused the trees to darken so the lightly coloured moths suddenly became very visible on the tree branches and got eaten more often.

In these areas, the melanistic form was less noticeable so it had higher rate of survival, and became more prevalent.



The switch from the light population to the dark population is an example of evolution as it shows how the species changes as a result of a changing environment by passing on its favourable genes.

### 5.1.2 Natural selection

Charles Darwin was the first scientist to publish the theory of natural selection, although Alfred Wallace had the same idea at the same time (but was just too slow to publish).

Natural selection suggests that the better adapted species will have a higher chance of survival and will therefore be able to pass on their genes and the species will evolve towards the better adapted species.

#### **Observations of natural selection**

#### There are more organisms than the environemtn can support

Individuals must compete for natural resources to survive. Different individuals of the same population have slightly different traits which make them better or less well adapted to the environment. Individuals with better adapted traits are more likely to catch pray, survive and breed, so their genes are passed on to the next generation. In this way, the favourable characteristics are increased within the population, and the *population gradually evolves*.

\* Note that only the genetically determined traits can be passed on this way. An athlete, whose muscles have grownn through exercise will not pass her big muscles to her offspring.

#### **Examples of natural selection**

#### Beak size of finches

Galapagos finches feed on seeds that fall on the ground, and for that, they have specially adapted beaks.

Some finches have larger, and some finches have smaller beaks. The larger beaks are better for eating larger, harder seeds.

During drought season, smaller seeds are not common, and only the large seeds are produced. If the drought extends for a long period of time, as it did between 1974 and 1977, the finches with larger beaks tend to be better adapted to cracking bigger seeds and are more likely to survive.



The mean beak size during those drought years was found have increased due to the fact that the animals with smaller beaks couldn't open the larger seeds and would therefore die before passing on their genes.

However, during a rainy season in 1983, there were more smaller seeds, and the animals with smaller beaks were faster to pick those up, so they had an advantage compared to the ones with larger beaks.

The mean beak size again decreased, as now the better adapted individuals were those with the smaller beaks, and they could breed and create more offspring.

#### Antibiotic resistance in bacteria

Bacterial antibiotic resistance is a growing problem in the world.

Normally, the antibiotic resistance gene exists in organisms that naturally produce the antibiotic.

This gene can be transferred to bacteria through the means of a plasmid.

The bacteria that have the gene cannot be killed by the antibiotic, so when it is administered to the patient suffering from a bacterial infection, all the non-resistant bacteria will die, and the resistant ones will survive and multiply.

Since these bacteria can't be killed by this type of an antibiotic, another antibiotic can be administered, killing this population, but giving a selective advantage to the ones possessing resistance gene for the other antibiotic.

In the end, only the resistant bacteria survive, multiply and exchange genes, leading to a broad range of ineffective antibiotics.

This can be avoided by limiting the antibiotic use, and by always taking the full does that ensures that all the non-resistant bacteria are killed and that they can't acquire resistance genes from the resistant bacteria.



# 5.2 Classification of biodiversity

## 5.2.1 Binomial system and dichotomous key

**Binomial system** is a universal system used to name the newly discovered species.

The first name in the system is *genus* and is written in the capital letter. The second name is the name of the species, written in the lower-case letter. When printed, the name is written in italics, such as *Homo sapiens*.

**Dichotomous key** is a tool that helps biologists distinguish between different organisms and classify them correctly.

An example of dichotomous key will be made after you learn about the classifications of plants and animals.

The key makes use of a series of statements about the features of an organism. These statements are numbered. Some statements lead to other statements that help better classify the organism. Other statements immediately give the name of the organism in question

**Classification** is the term for grouping of species.

**Taxonomists** are biologists that specialize in the field of classification.

Artificial classification refers to classifying organisms based on one feature that they share, but disregarding all the other ways in which they differ.

An example of this would be classifying all animals with a tail together. A squirrel and a lizard would both be classified together, even though they share very little in common, besides that, and their tails have different structure and function.

**Natural classification** is an alternative way of grouping organisms, based on their ancestry.

If species are grouped based on their common ancestor, they will share many more features. This ways of classification also helps identification of species, and is useful for prediction of their characteristics.



## 5.2.2 Domains and phylogeny

All organisms are classified into three main domains, namely *archea*, *eubacteria* and *eukaryota*.

Previously, the organisms were divided either into prokaryotes or eukaryotes, but archea and bacteria are as different from each other as they are from the eukaryotes.

Based on their RNA sequences, it seems that eubacteria and archea diverged very early in the evolution.

Viruses are not living organisms so they do not fit into any of the domains.



The 8 main taxa with the animal and plant exampls are:

Tuble 5.1. 6 main taxa					
Name	Human	Sequoia			
Domain	Eukaryota	Eukaryota			
Kingdom	Animalia	Plantae			
Phylum	Chordata	Coniferophyta			
Class	Mammalia	Pinopsida			
Order	Primate	Pinales			
Family	Hominidae	Cupressaceae			
Genus	Homo	Sequoia			
Species	sapiens	sempervirens			

Table 5.1: 8 main taxa



## 5.2.3 Classification of plants

Plants are classified into 4 main phyla, and you need to know how to distinguish between them, based on their structure.

#### **Bryophytes – Mosses**

- No real roots, but only rhizoids (root-like hairs)
- Simple leaves and stems
- No vascular tissues
- Reproduce using spores which are stored in capsules at the end of a stalk

#### Filicinophytes – Ferns

- Real roots and leaves, but short non-woody stems
- Leaves are often divided into pairs of leaflets
- Have vascular tissue
- Reproduce through spores which are made in sporangia, inside the leaves

#### **Coniferophytes – Conifers**

- Have real roots, leaves and woody stems
- Thick leaves with a waxy cuticle (imagine what a Christmas tree would look like)
- Have vascular tissue
- Reproduce using seeds which are made by female cones in structures called ovules
- Male cones will produce pollen used for fertilization

### Angyospermophytes – Flowering plants

- Real roots leaves and stems
- Some plants, like trees and shrubs have a woody stem, others don't
- Have vascular tissue
- Reproduce using flowers seeds are again made in ovules, but now in ovaries of flowers
- Fruits, which develop from these ovaries are used to disperse the seeds











## 5.2.4 Classification of animals

We will discuss 7 of the 30 possible animal phyla.

#### Porifera – Sponges

- No symmetry
- Attached to the surface
- No mouth or anus
- Contain pores



#### Cnidaria – Jellyfish

- Radial symmetry
- Tentacles and stinging cells
- Mouth, no anus



#### **Platyhelminths – Flatworm**

- Bilateral symmetry
- Flat body
- Unsegmented
- Mouth, no anus



#### **Annelida – Centipedes**

- Bilateral symmetry
- Segmented
- Mouth and anus





#### Arthropoda – Spiders and scorpios

- Bilateral symmetry
- Exoskeleton
- Segmented
- Appendages divided by joints



#### **Mollusca – Snails**

- Mostly bilaterally symmetrical
- Muscular foot and mantle
- Shell
- Mouth and anus



#### Chordata – Fish, mammals etc.

- Notochord and dorsal nerve chord (imagine a spine and nerves in it)
- Post-anal tail



# 5.2.5 Classifications of vertebrates

The phylum of chordata are very diverse, and we will now classify them further.

#### **Bony ray-finned fish**

Unlike in a dolphin with soft fins, these fish have fins supported by bones.

- Scales on skin
- Gills with one slit
- Fins supported by rays
- Swim bladder (so that they can change altitude in water)
- External fertilization



#### Amphibians (frogs)

- Soft, permeable skin
- Lungs with few folds
- External fertilization with gel that protects the eggs
- Larva live in water

#### **Reptiles (crocodiles)**

- Dry and scaly skin impermeable
- Lungs with a lot of folds
- Internal fertilization with soft shelled eggs
- One type of teeth

#### **Birds**

- Feathers on skin
- Lungs with parabronchi
- Wings, but no front legs
- Hard shells on eggs
- Beak, and no teeth

#### Mammals

- Hairs on skin
- Lungs with alveoli
- Give birth to live animals
- Mammary glands with milk
- Teeth of different kinds

Let's construct that dichotomous key, based on animal phyla!





# 5.3 Cladistics

Clade is a group of organisms that evolved from a common ancestor.

**Cladogram** is a tree diagram that shows how clades diverged one from another.

**Cladistics** is a method of classification that analyses base and amino acid sequence data to determine ancestry and construct cladograms.

Analogous traits are those traits that have a similar appearance and function, but have do not share the same origin.

Analogous traits arise from convergent evolution, meaning that species of different origins, developed similar features in response to their environment.

An example of this could be the wings of birds and bats, which have the same function even though the species developed these traits independently.

Homologous traits areare traits that are structurally different, but have the same origin.

Homologous traits are a result of divergent evolution, where a particular trait, shared by many species, accumulates structural differences as a response to the environment.

An example of this is the previously mentioned pentadactyl limb (first section of this chapter).

In the past, the only way to compare ancestry of certain species was to look at their anatomical features, but at some point, as in the case of bats and birds, it might be difficult to determine whether these features share a common ancestor, or have arisen by convergent evolution.

Today, cladistics takes advantage of possibility of genome sequencing. There is a positive correlation between the number of amino acid sequences, and the time since they have split from a common ancestor.



## 5.3.1 Human classification through cladistics

Mitochondrial DNA was used to classify humans and several other primates, using a cladogram. Mitochondria contain their own DNA, that is passed on from a mother to her child, and the size of this DNA molecules is much smaller than of a nuclear DNA molecule.

Still there are base pair differences that can help us determine the time points when certain primate species diverged from each other

- 5 million years ago: Human-Chimpanzee
- 140,000 years ago: African-European/Japanese
- 70,000 years ago: European-Japanese

## 5.3.2 Reclassification of figworts

Figworts were originally classified based on their structural differences. Sequencing of their genome showed that species within that family did not share a common ancestor.

Some plants were moved to pre-existing families (like plantain and broomrape). Others were transferred to completely new families based on evidence of a split between species. Some pre-existing families were found to actually share the ancestor with figworts, so these families were all merged into one.

With the developments in computers' analytical power and sequencing methods, there will probably be more and more re-classifications.

## 5.3.3 Cladogram analysis

Cladograms are constructed by plotting lines with branching points which represent points of divergence between species. These lines are usually plotted against time and percentage difference in the analysed variable (amino acid sequence, base pair sequence etc.).





#### **EVOLUTION AND BIODIVERSITY** | Cladistics



# HUMAN PHYSIOLOGY



## 6.1. Digestion and absorption

- Digestion: why is it essential? - Mixing and moving food along the gut: longitudinal and circular muscle contraction in the small intestine - Digestive enzymes - Absorption: the structure and function of villi

## 6.2. The blood system

- The cardiac cycle: blood circulation in the body - Control of heartbeat: the role of the pacemaker, brain signals and adrenalin

## 6.3. Defence against infectious 104 diseases - Blood clotting - Antibody production - HIV - Human

 Blood clotting – Antibody production – HIV – Human Immunodeficiency Virus

# 6.4. Gas exchange

- Ventilation: why do we need it? - Alveoli: the site of gas exchange - Inhalation and exhalation: muscle contractions needed to regulate airflow

## **6.5.** Neurons and synapses

- Action potential: generating and transmitting electrical signals in the brain - Neurotransmitters: chemical signalling across synapses



## 6.6. Hormones and homeostasis

Blood glucose – Diabetes – Thyroxine and metabolism
 Leptin and melatonin



# 116

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# 6.7. Reproduction

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 Embryo development – Steroid hormones – The menstrual cycle – In vitro fertilization – Harvey's reproduction experiments



# 6.1 Digestion and absorption

## 6.1.1 Digestion: why is it essential?

The food we eat consists of large and complex organic molecules, eg. *starch*, which *cannot be absorbed* by cells in the intestines. Digestion is the biochemical *breakdown of large, insoluble molecules into smaller ones*. For example, starch broken down into glucose is a useful source of energy. Excess glucose is stored as glycogen not starch since the latter cannot be traported through the bloodstream.

Figure 6.1: Digestive system.



Skill: Production of an annotated diagram of the digestive system (TIP: annotated suggests that the elements shown on the diagram must be briefly described, use the information on this section to correctly annotate the diagram of Figure 6.1 with the basic function of each labelled structure)



## 6.1.2 Mixing and moving food along the gut: longitudinal and circular muscle contraction in the small intestine

Partially digested food from the stomach enters the small intestine and moves down this structure due to *peristaltic sequences of muscle contraction*.

The small intestine has an inner layer of *longitudinal muscle* and an outer layer of *circular muscle*. Circular muscles contract behind the food to prevent backflow, whereas the longitudinal muscles contract to move the food along the intestine. When both layers of muscle contract simultaneously, they allow for the food to be mixed with digestive juices from the gall bladder and pancreas.

To enhance the absorption process, the surface of intestinal cells contain small *hair-like extensions* called *villi*, each of which contains a network of capillaries and a *lacteal* (a branch of the lymphatic system that enables lipid absorption) that connect to larger blood vessels and the lymphatic system. Figure 6.2: The structure of the small intestine.



## 6.1.3 Digestive enzymes

Skill: identification of tissue layers in transverse sections of the small intestine viewed with a microscope/micrograph

Physical breakdown of large food molecules is not enough, chemical breakdown by means of enzymes is thus necessary. Enzymes act as *biological catalysts* increasing the rate of digestion. This allows digestion to occur at *normal body temperature*. Several enzymes are needed as each is *substrate specific*. Note the source, substrate, products and optimal pH of each enzyme.

Enzyme type	Example	Source	Substrate	Products	Optimal pH
Amylase	Salivary amylase	Salivary glands	Starch	Maltose	7
Amylase	Alpha amylase	Pancreas	Starch	Maltose	7
Maltase	Intestinal maltase	Intestinal wall	Maltose	Glucose	7
Protease	Pepsin	Stomach wall	Proteins	Small polypeptides / amino acids	2-3
Endopeptidase	Trypsin	Pancreas	Proteins	Small polypeptides	7
Lipase	Pancreatic lipase	Pancreas	Triglycerides	Fatty acids + glycerol	7



## 6.1.4 Absorption: the structure and function of villi

The absorption phase is the process by which the products of digestion, mineral ions and vitamins are taken up through the villi that line the small intestine. To achieve maximal absorption the sturcture of the villus has a number of functional adaptations:

Villi have a large surface area to volume ration. They are one-cell thick structures which allows the products of digestion to easily pass through to the network of capillaries and a lacteal for quick nutrient absorption.



Figure 6.3: Structure of a villus

Each villi contains smaller structures called microvilli that further increase surface area for absorption of nutrients. These structures contain specific protein pumps and channels that also carry out active transport.

Absorption occurs via different processes, all of which have been discussed in previous chapters, depending on the type of molecule to be absorbed:

- Facilitated diffusion (e.g., hydrophilic nutrients like fructose)
- Simple Diffusion (e.g., hydrophobic nutrients like fatty acids)
- Endocytosis (e.g., larger molecules like cholesterol and triglycerides)
- Active transport (e.g., charged ions like calcium and sodium)

#### Digestion and absorption of starch derivatives

Starch consists of two different molecules: amylose and amylopectin, both linked by alpha-glucose 1,4 links. The only difference is that amylopectin also contains a few alpha-glucose 1,6 bonds. Amylase (salivary and pancreatic) can only break 1,4 bonds, digesting starch into maltose molecules and 1,6 bond-containing segments known as dextrins (which cannot be broken down by amylase). To further digest these



molecules, maltase and dextrinase in the small intestine convert the remaining molecules into glucose, which can then be absorbed by the villus through protein pumps (active transport).



Diagram showing the difference between amylose and amylopectin (two components of starch molecules). Amylose consists only of 1,4 bonds that result in a straight chain of glucose molecules. Amylopectin on the other hand contains "kinks" that result from 1,6 bonds between certain glucose molecules. These kinks cannot be broken down by amylase, and must be digested by dextrinase in the small intestine.

## 6.2 The blood system

# 6.2.1 The cardiac cycle: blood circulation in the body

In the 17<sup>th</sup> century, William Harvey proposed the theory of blood circulation that continues to be applied today. He demonstrated that the 4-chambered heart was the central "pumping mechanism" that caused blood to circulate the body at high pressures in arteries, and returned to the heart through veins. He also found that these two types of blood vessels are connected by small, hardly visible vessels now known as capillaries. Further research led him to conclude that blood vessels contain valves that prevent the backflow of blood, as well as to distinguish between two separate circulations that take place in the body:

**Pulmonary circulation** that carried deoxygenated blood from the heart to the lungs, where it becomes oxygenated and returns to the heart.





Systemic circulation that carries newly oxygenated blood to the remaining organs of the body, and returns deoxygenated blood back to the heart to enter pulmonary circulation.

Nowadays, the exact mechanism and structures involved in blood circulation has been clearly defined.

Figure 6.5 is a structure of our "central pump", labelled with the major blood vessels, valves and heart chambers involved in circulation.

The atria collect blood from veins (vena cava/pulmonary) at low pressure while the ventricles are contracting. Blood leaves the atria into ventricles ensuring the ventricles are full.

On the other hand, ventricles pump blood into arteries out of the heart. They can pump blood at high pressure because of their thicker, muscular walls. The heart valves work with the atria and ventricles to keep blood moving by preventing backflow. Note that the left ventricle supplies blood for the systemic circulation whereas the right ventricle supplies blood for pulmonary circulation.



Figure 6.5: Heart structure (cross section).



#### **Pulmonary circulation**

Deoxygenated blood from the superior and inferior vena cava collects within the right atrium.

) The walls of the right atrium contract, pushing blood from the atrium into the right ventricle through the atrioventricular valve.

Once a volume of blood has accumulated in the right ventricle, it contracts powerfully causing:

- right atrioventricular valve to close to prevent backflow;
- 2. increase of pressure in the right ventricle, leading to the opening of the right semilunar valve, pumping blood into the pulmonary artery.

Blood is carried by arteries, arterioles and capillaries in the lung alveoli, where it is oxygenated.

Venules and pulmonary veins return oxygenated blood to the left atrium.

#### Systemic circulation

Blood from the pulmonary veins collects in the left atrium.

The walls of the left atrium contract, pushing blood from the atrium into the left ventricle through the left atrioventricular valve.

Once a volume of blood has accumulated in the left ventricle, it contracts powerfully causing:

- 1. left atrioventricular valve to close to prevent backflow;
- increase of pressure in the left ventricle, leading to the opening of the left semilunar valve, pumping blood into the aorta.

The left ventricle has a much thicker musculature, as the aorta effectively distributes blood to the entire body, so a very powerful contraction is necessary

The aorta branches towards the entire body, one of the first branches directs blood to the coronary arteries (which supply the heart muscle with oxygenated blood for efficient muscle contraction). The rest of the blood is carried by arteries, arterioles and capillaries to various organs, to provide nutrients and oxygen.

Venules, veins and the inferior and superior vena cava return deoxygenated blood to the right atrium.



TIP: Try going through each circulation process using the diagram of the human heart on Figure 6.5! 1

2

3

#### HUMAN PHYSIOLOGY | The blood system

Both circulations occur simultaneously, so atrial and ventricular contractions occur in unison. Figure 6.6 is a graph of the pressure changes at each of the processes described previously in the atria and ventricles:





#### Coronary occlusion

Example.

Coronary artery occlusion, a common phenomenon caused by fatty plaques building up in the inner coronary arteries is a dangerous occurrence that restricts oxygen and nutrient supply to heart muscle, limiting contraction and thus blood circulation. This can cause chest pain and potential cardiac failure.

Some potential causes for this disease include:

- Hypertension
- Smoking
- High blood glucose (usually due to diabetes)
- High cholesterol levels
- Genetic factors

Figure 6.7: Coronary occlusion.





#### A comparison of the structure and functions of blood vessels

Artery	Capillary	Vein
Thick walls	Walls one-cell thick	Thin wall
No valves	No valves	Valves
High blood pressure	Low blood pressure	Low blood pressure
Carry blood from the heart	Link small arteries to small veins	Carry blood to the heart
Narrow lumen	Narrow lumen	Wide lumen

Skill: Identification of blood vessels as arteries, capillaries or veins from the structure of their walls.

Figure 6.8: Comparison of the structure of an artery, a vein and a capillary.



# 6.2.2 Control of heartbeat: the role of the pacemaker, brain signals and adrenalin

Cardiac muscle is *myogenic*, meaning that it can contract and relax without stimulation from the nervous system. Heart contrations are initiated at an in-built *pacemaker* (which keeps the cardiac muscle working in a coordinated sequence), called the *sinoatrial (SA) node*.

The SA node is a region of muscle cells in the right atrium that sets the basic pace of the heart the SA node produces the initial impulse that *causes both atria to contract at the beginning of each heartbeat*.

The *atrioventricular (AV) node* is also stimulated, and after a brief delay, stimulates *ventricular contration*, giving time for the ventricles to fill up with blood.

The natural rhythm of the pacemaker is modulated by the nervous system (signals from the medulla) and hormones (adrenalin).

Signals to speed up heart rate pass along the sympathetic nerve, and through the parasympathetic to slow down heart rate.

Emotions such as stress and increase in activity level cause the adrenal glands to release the hormone adrenalin (which stimulates the pacemaker to increase heart rate).



## 6.3 Defence against infectious diseases



# 6.3.1 Blood clotting

In case of a blood vessel injury, platelets and damaged cells release clotting factors.

These clotting factors cause the conversion of an inactive protein *prothrombin* into an active form called *thrombin*.

*Thrombin* further catalyses the conversion of insoluble *fibrinogen* to soluble *fibrin*, which is a long protein that forms a fibrous mesh that catches surrounding blood cells and forms a lump of blood called the blood clot.

If the clot is exposed to air, as is the case at the site of injury, it dries and protects the blood vessels from further entry of pathogens.

## **Blood clotting in coronary arteries**

In case of serious plaque deposits in coronary arteries (atherosclerosis), there is a high chance of the plaque rupturing and spilling into the blood stream.

The rupture, as well as the contact of blood with the plaque contents causes a clotting cascade to begin, leading to the formation of a blood clot.

Since coronary arteries are rather narrow, the clot often blocks the blood supply through this artery, and therefore the heart tissue supplied by this vessel stops receiving oxygen and nutrients.

If the supply is blocked for longer periods of time, this leads to the damage of heart tissue, termed heart attack, or to uncontrolled contractions of the heart called fibrillations.



Some heart attacks are less serious, and the heart can partially recover and start beating again, while more serious artery blocks lead to complete loss of heart function and death.

**Pathogen** is an organism that causes disease, for example a virus, bacterium or a fungus.

Recall that bacteria and fungi are live organisms, while viruses are not considered alive due to the lack of their own metabolic processes.

1<sup>st</sup> line of defence is the physical barrier, including the skin and mucous membranes.

Besides containing many tough layers, the *skin* is also provides a physical barrier in the form of acidic secretions that prevent the growth of pathogens on its surface.

*Mucous membranes* are parts of the skin covered in a secretion called mucous, that keeps the skin moist and prevents growth of bacteria by killing them with lysozyme enzymes

Mucous membranes can be found in the nose, throat, vagina and urethra.

 $2^{nd}$  line of defence is formed by blood cells inside our body.

*Phagocytes* are white blood cells that ingest pathogens through a process called phagoctyosis.

Once ingested, the pathogens are killed by the enzymes in cellular vesicles called lysosomes.

Phagocytes ingest pathogens in the blood and in other tissues, by leaving the capillaries and infiltrating the sites of infections.

Since phagocytes ingest any form of pathogens, they are said to form *non-specific immunity*.

*Specific immunity* is triggered by other types of white blood cells, called lymphocytes, which produce a response when in contact with a specific type of pathogen



## 6.3.2 Antibody production

Antigen is any sort of molecule that is recognised by our body as foreign.
Lymphocytes are white blood cells that are involved in specific imunity.
Upon encounter with antigens, lymphocytes can either activate other lymphocytes, or produce antibodies.
Antibody is a molecule produced by lymphocytes, in response to the recognition of antigens.

One type of lymphocyte can only produce one type of antibody, so our body contains many different types of lymphocytes.

Lymphocytes express this type of antibody on their surface so that an antigen can bind it and be recognised by our immune system.

The process of antibody formation is as follows:

- 1. Antigen binds a specific lymphocyte containing the matching antibody on its cell surface.
- 2. The binding of the antigen activates the lymphocyte which divides rapidly and produces many of the same antibody producing lymphocytes.
- 3. These new lymphocytes are called plasma cells because they produce large quantities of free antibodies which travel in the blood stream and bind the pathogen and mark it for destruction by other immune cells.
- 4. This is called specific immunity since the lymphocyte is specific for one type of pathogen containing a certain antigen.
- 5. Once the pathogen has been conquered, most of the lymphocytes die, while small fraction remains as memory cells which can quickly expand upon re-encounter with the same pathogen



## 6.3.3 HIV – Human Immunodeficiency Virus

HIV is a virus that reduces the effectiveness of the immune system by reducing the number of active lymphocytes.

HIV does so by penetrating the lymphocytes and its reverse transcriptase allows DNA to be produced from viral RNA.

This leads to a imparied ability to produce antibodies, thus the infected person has a lowered immunity and is more susceptible to infections.

Once our body has lost the majority of this sort of lymphocytes, the condition is termed AIDS, *acquired immunodeficiency syndrome*, since our body lacks the immunity formed by antibody production.

AIDS leads to death, as the body cannot fight even the most common infections such as the common cold.

Since HIV is a virus, it cannot survive long outside the body, so it is transmitted through certain bodily fluids:

- Through blood in hypodermic needles (often in drug abusers).
- Through unprotected vaginal, oral or anal sexual intercourse.
- Through the placenta or breast milk.
- Through transfused blood.

**Antibiotics** are chemicals produced by certain microorganisms which help the kill other invading organisms.

Penicillin is an antibiotic obtained from a Penicillium fungus which uses it to kill bacteria that might potentially invade it.

Antibiotics kill prokaryotic organisms (bacteria), and not eukaryotic (plant and animal), because they target metabolic processes specific for prokaryotes.

Therefore, antibiotics can kill bacteria in our body, without harming our own cells.

Viruses cannot be killed by antibiotics because they use the metabolism of the host cell, and therefore, to kill a virus, the cell containing it would also have to be killed.

With different types of antibiotics, most bacterial diseases can be contained, but due to the emergence of antibiotic resistance, some strains of bacteria cannot be so easily killed.


#### Florey and Chain

Florey and Chain tested penicillin on 8 mice that were infected with a pneumonia causing bacterium.

The treated mice recovered, while the untreated ones died.

Florey and Chain then treated the same antibiotic on patients dying from bacterial infections, and these patients recovered.

Today, drugs have to go through much more rigorous testing, before they can be given to humans.

First, the safety is thoroughly tested on animals.

Then, healthy humans are given the drug to assess if it is tolerated well.

If all of this goes well, a small number of very sick patients are given the drug, and if this is successful too, only then, a large scale study can be performed.



Example.

# 6.4 Gas exchange

# 6.4.1 Ventilation: why do we need it?

Ventilation is the process of bringing fresh air to the alveoli in the lungs and removing stale air (breathing in and out). This enables the process of gas exchange, where  $CO_2$  is removed from the alveoli and replaced by  $O_2$ .

Ventilation is essential because, in order for gas exchange to occur, a concentration gradient must be established (gas with higher  $O_2$  content must come into the lungs in order for it to diffuse into the alveoli, and  $CO_2$  to diffuse out).

Oxygen is essential for the processes like cell respiration.

Figure 6.9 is a diagram of the respiratory system, the flows through the trachea, bronchi and bronchioles until reaching the alveoli at the tips of the bronchioles, where gas exchange occurs between alveoli and pulmonary blood capillaries.



Figure 6.9: Ventilation system.



#### Lung cancer: causes and consequences

#### Causes:

Example.

- Smoking: tobacco smoke contains mutagens that can lead to tumour formation.
- Passive smoking: exhaled smoke from smokers passes carcinogens to others.
- Air pollution: e.g., nitrogen oxides from vehicles, diesel exhaust fuels.
- Radon gas.
- Asbestos and silica: dust from these materials causes cancer if deposited in the lungs.

Consequences:

- Difficulties with breathing.
- Chest pain.
- Persistent coughing.
- Loss of appetite.
- Weight loss.
- Coughing up blood.
- General fatigue.
- Can be fatal.

#### 6.4.2 Alveoli: the site of gas exchange

Alveoli are the body's gas exchange surfaces, formed in clusters at the ends of the smallest bronchioles. They are essentially air sacs with a very small diameter. The presence of many alveoli creates a large total surface area for gas exchange. Each alveolus is surrounded by a nextwork of blood capillaries allowing oxygen to diffuse through into the blood and carbon dioxide to diffuse in the opposite direction. Here are some additional structural characteristics that enhance this process:

• Two types of cells:

Pneumocytes type I extremely thin and adapted for gas exchange.

**Pneumocytes type II** secrete a solution containing surfactant, which creates a moist surface inside the alveoli to prevent the sides of the alveolus adhering to each other by reducing surface tension.

• Cells in alveoli are one-cell thick and in close proximity to blood capillaries: shorter diffusion distance for respiratory gases.



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Diagram showing type I and II pneumocytes, as well as the associated capillary. Direction of gas exchange is also shown.



Figure 6.10: Type I and II pneumocytes and capillary.

#### Emphysema: causes and consequences

Emphysema is a chronic and progressive respirtatory disease. Cilia that line the airways and expel mucus become damaged and cease to function appropriately, leading to a build-up of mucus that can lead to inflammatory response upon inhalation of smoke and air pollution. A protease is released from inflamed cells, causing the digestion of elastic fibres in the lung and eventually, alveolar walls collapse.

Causes:

Example.

- Smoking.
- Air pollution.

**Consequences:** 

- Loss of elasticity of the lungs.
- Reduced surface area for gas exchange.
- Difficult to exhale air → difficulty carrying out intense pH.



## 6.4.3 Inhalation and exhalation: muscle contractions needed to regulate airflow

#### **During Inhalation**

The external intercostal muscles contract while the internal intercostal muscles relax thus pulling the rib cage upwards.

The diaphragm contracts and flattens which increases the volume of thoracic cavity.

As a result of the reduction in pressure, air enters the lungs.

#### **During Exhalation**

In gentle exhalation, external intercostal muscles and diaphragm relax, reducing the volume of the thoracic cavity, increasing the pressure of the cavity and therefore releasing air out of the lungs. Forced exhalation also includes the contraction of internal intercostal muscles, further reducing the thoracic cavity volume.

Table 6.1

	Diaphragm	Internal intercostal	External intercostal	Volume	Pressure
Inhalation	Contracts	Relaxes	Contracts	High	Low
Exhalation	Relaxes	Contracts	Relaxes	Low	High

# 6.5 Neurons and synapses

# 6.5.1 Action potential: generating and transmitting electrical signals in the brain

In order for the brain to relay information to and from the rest of the body, neurons are the essential messengers that transmit electrical impulses to one another. Figure 6.11 is a diagram of a myelinated motor neuron, in charge of stimulating muscle fibres to contract.





The generation and transmission of electrical impulses across a neuron is achieved by an action potential.

**Resting potential** The charge difference across the membrane when a neuron is not firing (-70 mV), as maintained by the sodium-potassium pump.

Action potential The charge difference across the membrane when a neuron is firing (about 30 mV).

- 1. Before an action potential can be initiated, the neuron must first reach its resting potential (-70).
- 2. Resting potential is established the active pumping of K<sup>+</sup> ions into the neuron and of Na<sup>+</sup> ions out of it, creating concentration gradients for both ions, and an overall negative charge inside the neuron.
- 3. When input from a previous neuron is stimulating enough and the beginning of the axon reaches its threshold voltage (-55), an action potential is initiated.
- 4. Depolarization: Sodium voltage-dependent channels open and Na<sup>+</sup> diffuses into the neuron down its concentration gradient, reducing membrane potential and causing more sodium ions to open (membrane potential becomes more positive).
- 5. Repolarization: Potassium voltage-dependent channels open after a short delay due to the change in membrane potential, and K<sup>+</sup> ions diffuse out of the neuron down its concentration gradient. The inside of the neuron once again becomes more negative. In this stage, the electrical impulse has passed this section of the axon, and the sodium ions in the section adjacent open to transmit the impulse.
- 6. Concentration gradients of Na<sup>+</sup> and K<sup>+</sup> across the membrane are restored by the Na<sup>+</sup>/K<sup>+</sup> pump, and the membrane in this section of the axon once again reaches resting potential (and is once again ready for another action potential).

In myelinated neurons (as seen in Figure 6.11), this ion exchange only occurs at the Nodes of Ranvier, thus conduction of the action potential "jumps" down the axon far more quickly than in unmyelinated axons, where numerous adjacent sections of the axon must undergo the process outlined previously to carry the electrical impulse down the length of the axon. This is known as "saltatory conduction".



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Action potential graph showing the processes of depolarization, repolarization and the re-establishing of membrane potential.

# 6.5.2 Neurotransmitters: chemical signalling across synapses

Once an action potential has reached the axon terminals, chemical signals are necessary to pass on information from the presynaptic neuron to the postsynaptic one or onto an effector cell (e.g., muscle fibre). Separating each neuron are small junctions known as synapses, through which specific chemical messengers called neurotransmitters (NTs) can diffuse. An overview of this process is:

- 1. A nerve impulse reaches the axon terminal of the presynaptic neuron, leading to depolarization at the terminal.
- 2. Depolarization leads to the opening of calcium channels and an influx of Ca2+ into the presynaptic neuron.
- 3. Influx of calcium causes neurotransmitter-filled vesicles to fuse with the presynaptic membrane and release NTs into the synaptic cleft by exocytosis.
- 4. NTs diffuse across the synaptic cleft and bind to specific NT receptors in the postsynaptic neuron.
- 5. This binding results in the opening of ligand-gated ion channels that allow either Na<sup>+</sup> or Cl<sup>-</sup> to diffuse into the postsynaptic neuron.
  - (a) Na<sup>+</sup> influx creates an excitatory signal at the postsynaptic membrane, and allows for the transmission initiation of the action potential if the threshold potential is reached.
  - (b) Cl<sup>-</sup> influx on the other hand, leads to hyperpolarization (inside of the neuron becomes more negative), resulting in an inhibitory signal that may prevent action potential initiation.



6. NTs in the synaptic cleft are rapidly degraded by enzymes or are taken up by the presynaptic neuron once more. Ca2+ is pumped out of the presynaptic neuron as well to re-establish a concentration gradient

#### Cholinergic synapses

A common example of a synapse is the cholinergic synapse, which most commonly results in the opening of  $Na^+$  ion channels in the postsynaptic membrane and thus transmits an excitatory signal. Acetylcholine is the NT that is released in cholinergic synapses, which then diffuses and binds to nicotinic receptors at the postynaptic neuron. Enzyme cholinesterase rapidly degrades this NT where one of the residual components, choline, is reabsorbed into the presynaptic neuron.

Some chemicals found in neonicotinoid pesticides have a similar chemical structure as acetylcholine, and can bind to the acetylcholine receptors in cholinergic synapses. Cholinesterase is unable to degrade this chemical and thus, the cholinergic receptor remains blocked and nerve impulses cannot be mediated. This is a very dangerous phenomenon that is lethal for bees and insects that come into contact with these types of pesticides.

# 6.6 Hormones and homeostasis

## 6.6.1 Blood glucose

Glucose concentration in blood has to be well tightly regulated in order to provide cells with enough energy, but at the same time, to control water movement in and out of the cells.

Pancreas is the organ in charge of glucose regulation, and it produces two hormones:

Glucagon is produced by *alpha cells* and is secreted when the glucose levels are low:

- it leads to breakdown of glycogen and its release into the glood;
- this happens in the liver.

Insulin is produced by *beta cells* and is secreted in response to high blood glucose levels:

- it stimulates cells to take up the glucose from blood;
- it also stimulates liver and muscle cells to store glucose in the form of glycogen.



Example

## 6.6.2 Diabetes

Diabetes mellitus is a disease of faulty glucose regulation. Two types of diabetes can be differentiated, based on the cause.

#### **Type I diabetes** Type II diabetes early-onset diabetes usually develops in adult-onset diabetes usually occurs in those under 20 years old those over 40 years old inability to produce sufficient inability to respond to insulin mainly quantities of insulin due to insufficient receptors on target cells target cells remain sensitive to insulin target cells less sensitive to insulin is linked with is linked with • dietary genetic predisposition • virus • lifestyle factors • autoimmune disorder increased fatty acids in blood • destruction of (pancreatic) beta cells involved • controlled by diet requires daily injections of insulin • beta cell transplant • exercise

- weight loss
- medication but not insulin

#### injections

# 6.6.3 Thyroxine and metabolism

**Thyroxin** is a hormone produced by the thyroid gland which is responsible for the metabolic activity of the cells.

Thyroxin is made out of 4 molecules of iodine. Low iodine intake can result in lack of thyroxin synthesis (which is why all the salt is additionally iodized).



Thyroxin causes an increase in metabolic rate of the cells, leading to higher heat production by the body. It also causes vasoconstriction, shivering and heat production by brown adipose tissue.

When the body temperature drops, this is sensed by the hypothalamus in the brain which triggers the production of thyroxin by the thyroid.

Thyroxin increases the metabolic rate of the cells, causing higher heat productions, but also leads to vasoconstriction of skin blood vessels (to preserve heat) and shivering.

# 6.6.4 Leptin and melatonin

**Leptin** is a hormone, first discovered in mice and later in humans, secreted by adipose (fat tissue) cells which controls appetite.

Leptin is produced by the adipose cells and travels to the hypothalamus in the brain, where it stimulates certain cells to signal decrease of appetite.

In such a way, the body is able to regulate its body mass, by decreasing food intake, with increased fat tissue generation.

The hormone was then used in experiments on obese mice lacking the gene for leptin, to see if it could restore their normal body weight.

Since the mice lost weight after leptin administration, a similar trial was started in humans.

However, it turnet out that incjections of leptin did not cause a significant decrease in body weight in obese humans.

This was explained by the observation that the body produces sufficient leptin, but the hypothalamus cells became insensitive to it.

Only a small number of obesity cases in humans is due to lack of leptin production by the body.

**Melatonin** is a hormone produced by the pineal gland and it is responsible for controlling the sleep-wake cycles in humans.

Cells in the eye send impulses to the supra chiasmatic nuclei (SCN) in the hypothalamus about the amount of light/dark.



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SCN controls melatonin production, by increasing it at night, and decreasing it at dawn.

Over the years, out melatonin production decreases so our sleep cycles become less regular.

When traveling long distances by plane, SCN and pineal gland cannot adjust to the new time zone rapidly enough, so during the first few days, traveller's body continues functioning by the old rhythm. This is termed *jet lag*. Taken oral doses of melatonin at the newly designated sleeping times can help the body adjust to the new time zone more rapidly.

# 6.7 Reproduction



Figure 6.13: Female reproductive system.

Figure 6.14: Male reproductive system.



ACADEMY

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# 6.7.1 Embryo development

The difference between a male and a female zygote is in the 23 pair of chromosomes.

A male has an XY combination, while the female has an XX combination.

A gene called SRY is present in male zygotes, but not in female ones.

If the SRY gene is present, it codes for a testis determining factor protein which causes development of testes.

## 6.7.2 Steroid hormones

- **Testosterone** is produced by testes, both in a foetus and later in life. In a a foetus, testosterone leads to development of male genitalia. During puberty, it leads to development of secondary sex characteristics (growth of pubic hair, penis and testes). In later life, testosterone stimulates sperm production.
- **Oestrogen** is produced in the ovaries. In foetus, it causes development of female genitalia (if there is no testosterone). In puberty, it leads to development of secondary sex characteristics.
- **Progesterone** is also produced in the ovaries and its function is to thicken the uterus before embryo implantation.

**Negative feedback** means that the increase in a product of a process will lead to the decrease in the process itself.

**Positive feedback** means that the production of the final product will stimulate the process of production even further.



# 6.7.3 The menstrual cycle

In order to fully understand the processes during the menstrual cycle, the following terms should be clearly explained.

(••)
<b>Pituitary hormones</b> are hormones produced in the pituitary gland (FSH and LH).
<b>FSH</b> stimulates follicle development and secretion of oestrogen by the follicle.
<b>LH secretion</b> is caused by high oestrogen levels and its main function is to cause the release of the egg and maintain the development of the follicle wall after ovulation.
<b>Ovarian hormones</b> are hormones produced in the ovaries (oestrogen and progesterone).
Oestrogen is produced in a positive feedback with FSH by the follicle wall, it causes repari and thickening of the uterus lining and causes LH secretion.
<b>Progesterone</b> is produced by the corpus luteum and maintains thickening of the uterus.
Together, these two hormones inhibit LH and FSH hormones.

The aim of the menstrual cycle is to mature and release an egg, ready for fertilization, and to prepare the uterus for embedding the embryo in its wall.

- 1. The first, day 0 of the menstrual cycle, is the first day of bleeding.
  - (a) FSH increases and stimulates the follicles with the egg to develop.
  - (b) The development of follicles leads to oestrogen secretion.
  - (c) Oestrogen and FSH are in positive feedback because oestrogen production makes follicle cells more sensitive to FSH, which in return results in more oestrogen production.
  - (d) In this way, both oestrogen and FSH increase.
  - (e) Oestrogen leads to repair of the uterus lining.



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- 2. Around day 14, the uterus has gotten thicker, and the follicular phase of development is ending.
  - (a) When oestrogen is high enough, it causes LH production by the pituitary gland.
  - (b) At the same time, peaks of oestrogen lead to FSH inhibition, which in turn then leads to decrease in oestrogen (negative feedback).
  - (c) As LH reaches a peak, it causes completion of meiosis in the egg and release of the egg into the oviduct-ovulation (around day 14).
- 3. After ovulation, the broken follicle maintains the uterine lining, in case fertilization occurs.
  - (a) LH stimulates the burst follicle to develop into corpus luteum.
  - (b) Corpus luteum produces progesterone, and some oestrogen.
  - (c) Progesterone will continue rising for a few days in order to maintain the thickening of the uterine lining.
  - (d) The increase of progesterone and oestrogen levels inhibits the FSH and LH production by the pituitary (thereby preventing another ovulation).
- 4. If no fertilization occurs, the corpus luteum starts degenerating, and the menstruation will begin.
  - (a) With degeneration of corpus luteum, progesterone and oestrogen levels start falling, therey lifting the inhibition of FSH.
  - (b) The drop in progesterone results in degeneration of the uterine lining and therefore, menstrual bleeding.
  - (c) As FSH levels can begin to rise again, a new cycle of follicular development can start.

# 6.7.4 In vitro fertilization

**In vitro fertilization (IVF)** is a procedure of fertilization that is done outside of the female body, in order to overcome the fertility issues of either partner.

- 1. Drugs are used to down-regulate the menstrual cycle.
- 2. FSH injected to stimulate many follicles to develop.
- 3. HCG injected to cause the follicles to mature.
- 4. The eggs are harvested from the ovaries and the semen sample is collected.



- 5. The is semen mixed with eggs in a dish outside the body to allow for fertilization.
- 6. The dish is incubated at 37 °C allowing the embryos to develop sufficiently for implantation.
- 7. The dish examined to choose healthiest embryo.
- 8. The embryos are placed in uterus using a catheter. One, two, three and up to four (in some countries) embryos may be implanted.
- 9. Finally a pregnancy test used to see if procedure has been successful.

Used in cases of

• blocked oviduct;

• infertility;

• low sperm count;

• need for genetic screening;

cannot become pregnant; need for donor embryo.

# 6.7.5 Harvey's reproduction experiments

William Harvey was a scientist in the 17 century who wanted to explain how copulation resulted in offspring. He chose to study deer during their mating season. He would slaughter the deer after the mating, in order to observe the development of offspring in the uterus.

The current theory suggested that the male produces an egg that is activated by menstrual blood, and then develops into a foetus in the human uterus.

Harvey showed that this theory was wrong, but he couldn't show that the foetus development was a result of copulation, since the developing embryo was too small to see without a strong microscope.

The story of William Harvey shows how science can only progress with a simultaneous progress in the apparatus and scientific techniques.



HUMAN PHYSIOLOGY | Reproduction



# **NUCLEIC ACIDS**



7.1. DNA structure and replication

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– Nucleosomes: bundles of DNA and protein – DNA replication – Non-coding DNA

# 7.2. Transcription and gene 128

#### expression

Transcription – Transcription regulation by nucleosomes
 Post-transcriptional mRNA modifications – Gene
 expression: regulation by protein binding and environmental factors

# 7.3. Translation

– Translation – Free vs bound ribosomes – The 4 levels of protein structure



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# 7.1 DNA structure and replication

# 7.1.1 Nucleosomes: bundles of DNA and protein

Nucleosomes are the units into which chromatin is organized in the eukaryotic nucleus. One nucleosome consists a globular structure, with a core of 8 histone proteins with DNA wrapped around them. Histone H1 links DNA to the protein core, and each "bundle" of DNA is linked to the next by linker histones. The N-terminal tails of the histones extend outwards from the core of the nucleosome and can attach to neighbouring nucleosomes, pulling them closer together and enabling these to supercoil during the condensation of chromosomes in mitosis. Additional protein modification to the histones (e.g. methylation and acetylation) also affects their level of condensation (for instance, if a methyl group is added, nucleosomes cannot be as closely packed). This serves to influence processes like trancsription, by making the DNA available for enzymatic action, or supercoiling to prevent enzyme access. Molecular visualization software van be used to analyse the association of protein and DNA in nucleosomes.

Figure 7.1: Structure of a nucleosome



# 7.1.2 DNA replication

Having established the detailed structure of DNA, which contains antiparallel strands that each consist of a sugar-phosphate backbone and one of 4 nitrogenous bases, that attach to the antiparallel strand by the formation of hydrogen bonds. These bases are:

- Adenine (purine) forms two hydrogen bonds with Thymine (pyrimidine)
- Guanine (purine) forms three hydrogen bonds with Cytosine (pyrimidine)



This very specific binding scheme led to a suggested mechanism for DNA replication which is semi-conservative:

- DNA replication occurs in a 5'-3' direction, as DNA polumerases can only add free nucleotides to the 3' end of a primer
- Since the double stranded helix is unwound during replication, each "parent" strand serves as a template dor the newly forming "daughter" strand → one parent strand is always conserved in newly formed DNA molecules
- The binding specificity reduces the risk of mutations, as each nigtrogenous base has a high binding affinity for its complementary base, and a lower one for the two remaining bases.

Below is an explanation of the replication process in prokaryotes at the two parent strands, also known as leading strand (where replication occurs continuously from the end of chromosome towards replication fork) and the lagging strand (where replication is not continuous and occurs from the replication fork towards the end of the chromosome):

- 1. Helicase unwinds and separates the DNA double-helix into the two parent strands, and DNA gyrase binds at the replication fork to stabilize the DNA during this process
- 2. Single strand binding proteins bind to the resulting parent strands to stabilize them and prevent them from reattaching and coiling once more

#### At lagging strand (3' - 5' direction)

- 1. Primase attaches RNA primer is attached to begin replication at the site nearest to the fork, which contains the 3' end of the parent strand
- 2. As DNA continues to unwind and separate, more primers are attached at the replication fork
- 3. DNA polymerase III adds all the free nucleotides in a 5' 3' direction
- 4. DNA polymerase I replaces the RNA primer with DNA nucleotides
- 5. The separate segments that form between each primer Okazaki fragments
- 6. Enzyme DNA ligase joins Okazaki fragments together

#### At leading strand (5' - 3' direction)

- 1. Primase attaches RNA primer is attached to begin replication at the 3' end of the leading strand
- 2. DNA polymerase III adds all the free nucleotides in a 5' 3' direction
- 3. DNA polymerase I replaces the RNA primer with DNA nucleotides



# 7.1.3 Non-coding DNA

It is important to realize that not all of the DNA present in the cell nucleus is later transcribed and translated to form proteins. Some special cases of non-coding DNA have very important functions in and outside the nucleus:

- Regulating gene expression: sites where proteins can bind to promote or repress transcription of specific genes
- Introns: in eukaryotes, these are non-coding sequences that "interrupt" a coding sequence, and are important in mRNA processing
- Telomeres: sequences located on tht tips of the chromosomes. After each replication event, a small segment at one end of the chromosome cannot be replicated, and that part of DNA is lost, thus leading to the shortening of telomeres (this way, important genese are not immediately lost)
- Genes for tRNAs and rRnas: while these genes do not code for proteins, they do code for essential machinery needed during translation

# 7.2 Transcription and gene expression

# 7.2.1 Transcription

As in DNA replication, the process of transcription, where DNA is transcribed into mRNA that can exit the nucleus and be translated by ribosomes, occurs in a 5'-3' direction. RNA polymerase can only add free RNA nucleotides to the 3' end of the growing mRNA molecule. Below is the description of the processes involved in transcription:

- **Initiation:** RNA polymerase binds to the promoter region of the gene (this region contains a specific base sequence and signals the enzyme to initiate transcription)
- Elongation: RNA polymerase simultaneously uncoils and transcribes the coding region in a 5'-3' direction
- **Termination:** RNA polymerase detaches from the mRNA strand after reaching the terminating sequence, the mRNA then "peels" away from the DNA template away from the DNA template and moves to be modified and later translated outside the nucleus

The DNA strand that is translated is called the "antisense" strand (the mRNA of interest will contain the sequence that is identical to the "sense" strand, which codes for gene of interest, with uracil instead of thymine).



# 7.2.2 Transcription regulation by nucleosomes

As mentioned earlier, the coiling and uncoiling of DNA by nucleosome interactions can regulate the process of translation. DNA regions of highly condensed chromatin will usually not be transcribed, as enzymes cannot easily access the DNA (regions that remain highly condensed at all times are known as heterochromatin, and are normally not transcribed). On the other hand, decondensed chromatin is easily accessible for transcription enzymes and transcription factors, and is readily transcribed.

## 7.2.3 Post-transcriptional mRNA modifications

In eukaryotic organisms, mRNA is modified before leaving the nucleus to be translated. The major modification consists in the removel of introns by RNA splicing, which "cuts out": the non-coding sequences and joins the remaining exons together. Many genes have several exons that can be spliced in various ways to create a large range of different proteins.

# 7.2.4 Gene expression: regulation by protein binding and environmental factors

The process of transcription can be regulated by both proteins and environmental factors. Proteins that bind to the promoter region, known as transcription factors, are an example of this, as they enhance RNA polymerase binding and promote the transcription of specific genes. Environmental factors can also affect gene expression in a way that is not inheritable. The study of epigenetics focuses on these factors and the way they affect this process, by altering transcription, mRNA modifications, translation, etc. For example, diet, age, diseases can influence changes in gene expression that do not necessarily involve direct alteration of the DNA sequence. Some epigenetic modifications include:

- DNA methylation: can fix genes in an "off" position, preventing transcription
- Histone modification: acetylation and methylation can affect the level of chromatin condensation
- microRNAs: can bind to mRNA, leading to the degradation of the entire complex
   → gene cannot be translated



# 7.3 Translation

# 7.3.1 Translation

**Translation** is the process in which gene transcripts are converted into large polypeptide chains that can then be folded and modified to create functional proteins.

#### Initiation

The tRNA molecule with the start anticodon UAC binds to the small subunit of the ribosome (peptide chain site on diagram).

The small subunit then binds the 5' end of the mRNA strand and slides along in a 5'-3' direction until it finds the start codon on the mRNA (AUG).

The large and small subunit of the ribosome then bind together.

A tRNA molecule with the anticodon complementary to the next mRNA codon on the chain binds to the amino acid site of the ribosome.

The elongation process begins.

#### **Elongation**

The anticodon of incoming tRNA pairs with its complementary mRNA codon at the amino acid site of the ribosome.

The amino acids in the tRNAs at the peptide and amino acid sites form a peptide bond, and the growing polypeptide chain remains attached to the tRNA at the amino acid site.

#### **Translocation**

The large subunit od the ribosome moves forward and the small unit slides after it, moving the entire ribosome 3 nucleotides down the mRNA strand.

The tRNA in the peptide site moves to the exit site (where the tRNA is released and can attach to a new amino acid).



The tRNA carrying the growing polypeptide chain now moves to the peptide site.

The next codon to be translated is at the amino acid site, where its complementary tRNA codon can bind.

The process repeats itself.

#### **Termination**

When one of three stop codons (UAG, UAA, UGA) reaches the amino acid site of the ribosome, the large subunit advances over the small subunit, detaching from it and causing the polypeptide chain to be released from the last tRNA.

The polypeptide can then fold and be further modified to create a functional protein.

It is important to realize that translation of a single mRNA sstrand can occur in multiple ribosomes (many can attach at different sites), creating a polysome. Polysomes can be visualized using molecular visualization software.

Figure 7.2: tRNA & ribosome structure



It is important to realize that each tRNA molecule can only bind a specific amino acid, depending on its anticodon (triplet of bases located on the middle loop of the tRNA).

The amino acid binds at the 3' end of the single stranded molecule

20 different amino acids, some of which can bind to several tRNAs (same amino acid, different anticodons)



This binding is catalysed by tRNA-activating enzymes, which use the principle of substrate-enzyme specificity to attach the correct amino acid the appropriate tRNA molecule





# 7.3.2 Free vs bound ribosomes

In prokaryotic organisms, free ribosomes can begin the process of translation immediately after transcription, as there is no nuclear membrane that can inhibit ribosome action in these organisms. In eukaryotes however, the mRNA must first leave the nucleus through nuclear pores and can then be translated at either free cytoplasmic ribosomes, or bound ribosomes attached to the rough endoplasmic reticulum.

Free ribosomes release the protein into the cytoplasm, and the proteins formed by these organelles are mostly for use inside the cell (e.g. transcription factors, intracellular enzymes, etc.)

Bound ribosomes release the polypeptide chain into the rough endoplasmic reticulum, where the protein is modified, folded and packaged into vesicles that can then be secreted out of the cell or taken up by lysosomes.

# 7.3.3 The 4 levels of protein structure

So far we have looked some of the essential steps in gene expression. The genes are first transcribed from their DNA sequence. The resulting mRNA strand is modified and moves out of the nucleus, where free or bound ribosomes translate it into large polypeptide chains, that upon folding and modification become functional proteins. Below is an overview of the four levels of protein structure.

#### **Primary structure**

The primary level of protein organization consists of the unique sequence of amino acids held together by peptide bonds that form a polypeptide.



Skill: Identification of polysomes in electron micrographs of prokaryotes and eukaryotes → In the diagrams above, you can clearly see the thin mRNA strand with multiple ribosomes attached to it, each translating the same mRNA (the darker circles show the growing polypeptide chains at each ribosome) The amino acid sequence can consist of any combination of the 20 existing amino acids, in any given order.

The sequence is determined by the organism's DNA.

Primary structure determines the function, structure and location of the protein (small alterations here can lead to major changes in the resulting protein).

#### Secondary structure

The secondary structure results from the interaction between the single polypeptide chain, and can result in two different structures:

- 1.  $\alpha$ -helix: the polypeptide is wound into a right-handed helix when hydrogen bonds are formed between adjacent turns of the helix (bonds between N–H and H–O from neighbouring amino acids).
- 2.  $\beta$ -pleated sheets: polypeptide chains create parallel folds forming hydrogen bonds between them (flat rather than helical).

#### **Tetriary structure**

The polypeptide chain bends and folds over itself because of the interactions of the separate amino acids' R-groups and the peptide backbone. Some interactions that occur are:

- Covalent bonds between sulphur atoms form strong disulphide bridges.
- Hydrogen bonds between polar side chains.
- Van der Waals interactions.
- Ionic bonds between positively and negatively charged side chains.

#### **Quaternary structure**

Multiple polyeptide chains combine to form a single protein. Not all proteins do so. Conjugated proteins bind a prosthetic or non-polypeptide group (e.g. haemoglobin).



#### NUCLEIC ACIDS | Translation



# METABOLISM, CELL RESPIRATION AND PHOTOSYNTHESIS

# 8.1. Metabolism A Metabolic pathways: chains and cycles - Enzymes & activation energy of reactions - Competitive vs non-competitive inhibition - End-product inhibition 8.2. Cell respiration: a redox reaction - Glycolysis: from glucose to pyruvate - Link reaction and Krebs cycle: aeorobic cell respiration - Electron transport chain and chemiosmosis: ATP - Mitochondria: site for cell respiration

8.3. Photosynthesis

Light-dependent reactions – Light-independent reactionsChloroplasts: the site for photosynthesis

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# 8.1 Metabolism

## 8.1.1 Metabolic pathways: chains and cycles

Metabolic pathways are the specific sequences in which reactions occur; at each step, an enzyme catalyses each reaction.

Some metabolic pathways consist of chains of reactions (e.g. glycolysis), where the initial substrate and end product do not necessarily interact or resemble one another. Other metabolic pathways consist of cycles of reactions (e.g. Krebs cycle), where a substrate of the cycle is constantly regenerated from other intermediated involved in the same cycle. Many metabolic pathways include both kinds of reactions.



Figure 8.1: Diagram showing a chain and a cycle of reactions

# 8.1.2 Enzymes & activation energy of reactions

Enzymes are globular proteins that function as biological catalysts that speed up chemical reactions in cells by binding substrates to their active site. The way enzymes catalyse reactions is as follows:

Before a molecule of the reactant can take part in the reaction, it has to gain energy (also known as activation energy), which helps break the molecular bonds within the reactant.

When new bonds are made after the reactant is broken down, energy is released (see graph below).



Enzymes reduce the activation energy of the reactions they catalyse by weakingn the reactant's molecular bonds, making it easier for the reaction to occur.



Graph showing the reduction in activation energy in the presence of enzymes, as well as the fact that the energy released by the formation of new bonds remains unchanged. The reaction shown above is exothermic, meaning that it releases more energy than its activation energy (opposite is endothermic)

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# 8.1.3 Competitive vs non-competitive inhibition

#### **Competitive inhibition**

A competitive inhibitor competes directly for the active site of the enzyme by binding to it. It prevents the substrate from binding until the inhibitor dissociates. The inhibitor must have a very similar molecular structure as the substrate in order to fit the active site and be able to bind to it.

Can be reversible or irreversible.

Increasing substrate concentration can usually reduce the inhibitory effect, as the inhibitor is more readily displaced by the many substrate molecules.

In the Krebs cycle, succinate dehydrogenase acts on substrate succinate, removing a hydrogen atom and converting it itno another intermediate.

Malonate acts as a competitive inhibitor, effectively slowing down the Krebs cycle.





#### **Non-competitive inhibition**

A non-competitive inhibitor binds to the enzyme at a different site from the active site (also known as allosteric site).

Binding of the inhibitor at the allosteric site causes a conformational change in the enzyme (active site included), making it non-functional and slowing down the reaction.

Usually reversible (not always), but unlike competitive inhibition, increasing substrate concentration does not reduce the inhibitory effect (even if there are more substrate molecules, they cannot bind to the non-functional active site).

# Nerve gas (Sarin) binds to respiratory enzyme acetyl cholinesterase allosterically, changing the shape of the enzyme's active site.

Substrate can no longer bind, and this disruption can result in a blockade of the respiratory pathway  $\rightarrow$  can be lethal, as this process is irreversible



#### 8.1.4 End-product inhibition

End-product inhibition occurs when the product of the last reaction in a metabolic pathway (chain or cycle) inhibits the enzyme that catalyzes the first reaction.

The inhibitet enzyme is called the allosteric enzyme. Allosteric enzymes have both an active site and an allosteric site.

The end product binds to the allosteric site and changes the shape of the active site, inhibiting the enzyme, this process is reversible, so when the end product detaches, the reaction continues. Efficient way of controlling metabolic rate. If the concentration of the product is too high, end-product inhibition stops the pathway, producing less waste.



Skill: Distinguish between different types of inhibition from graphs at specified substrate concentration. <u>Example.</u>

# 8.2 Cell respiration

#### 8.2.1 Cell respiration: a redox reaction

Cell respiration is the controlled release of energy, in the form of ATP, from organic compounds in cells.

Cell respiration is a redox reaction, which involves the oxidation and reduction of electron carriers NAD<sup>+</sup> and FAD at different stages of this metabolic process (oxidation is the loss of electrons and hydrogen; whereas reduction is the gain of electrons and hydrogen).

Oxidation	Reduction	
Loss of electrons	Gain of electrons	
Gain of oxygen	Loss of oxygen	
Loss of hydrogen	Gain of hydrogen	
Results in many C—O bonds	Results in many C–H bonds	
Results in a compound with lower	Results in a compound with higher	
potential energy	potential energy	

#### Table 8.1

## 8.2.2 Glycolysis: from glucose to pyruvate

Glycolysis is a chain reaction pathway that is catalysed by enzymes in the cytoplasm of the cell and converts a glucose molecule into two pyruvates. It uses no oxygen, so it happens in aerobic and anaerobic cell respiration (in both prokaryotes and eukaryotes). It is divided in four main phases:

#### Activation / Phosphorylation phase

Two molecules of ATP are used to begin glycolysis. One phosphate from each ATP phosphorylates the glucose molecule to form intermediate molecule fructose bisphosphate

#### Lysis phase

This 6-carbon intermediate is split into two 3-carbon sugars, each containing one phosphate group

#### Oxidation phase

Two hydrogen atoms are removed from each 3-carbon molecule (oxidation). The energy released by this oxidation is used to link another free phosphate group to



each 3-carbon molecule. The hydrogen atoms are then carried by electron carrier NAD<sup>+</sup> (forming NADH+H<sup>+</sup>)

#### ATP formation phase

Pyruvate is formed by removing the two phosphate groups from each 3-carbom molecules and phosphorylating ADP (yielding 4 ATP molecules). Because 2 ATP were initially used in the activation phase, glycolysis yields the following net products:

- $2 \times ATP$
- 2 × pyruvate
- $2 \times \text{NADH} + \text{H}^+$





# 8.2.3 Link reaction and Krebs cycle: aeorobic cell respiration

If oxygen is readily available to the cell, the pyruvate molecules undergo the link reaction and the Krebs cycle to create a much higher yield of ATP. Below is a description of both processes:

#### **Link reaction**

Pyruvate enters the mitochondrion's matrix through active transport. In the matrix, enzymes remove hydrogen (oxidation) and carbon dioxide (decarboxylation) from each pyruvate molecule. This is called oxidative decarboxylation. Hydrogen is carried away by NAD<sup>+</sup>. The resulting 2-carbon molecule binds to coenzyme A (CoA), forming end product acetyl-CoA

Link reaction yields:

- $2 \times CO_2$
- $2 \times \text{NADH} + \text{H}^+$
- $2 \times acetyl-CoA$

#### **Krebs cycle**

Occurs in the matrix of the mitochondrion.

Acetyl-CoA combines with a 4-carbon compound, forming a 6-carbon intermediate (all the products mentioned below double, since there are two acetyl-CoA molecules that enter the cycle).

The 6-carbon intermediate undergoes oxidative decarboxylation, resulting in a 5-carbon molecule,  $CO_2$  and NADH+H<sup>+</sup>.

Another oxidative decarboxylation occurs, resulting in a 4-carbon molecule,  $CO_2$  and NADH+H<sup>+</sup>.

The 4-carbon intermediate undergoes substrate-level phosphorylation, releasing a phophate group to phosphorylate an ADP molecule. The resulting intermediate is then oxidized, this time by  $FAD \rightarrow FADH_2$ .

The resulting 4-carbon molecule is the same as the starting intermediate that combines with acetyl-CoA at the beginning of the cycle.



Overall, the two acetyl-CoA molecules lead to the following products:



CO<sub>2</sub> is released as a waste product (in total 6 molecules).

# 8.2.4 Electron transport chain and chemiosmosis: ATP

NADH<sup>+</sup>H<sup>+</sup> and FADH<sub>2</sub> carry hydrogen and electrons (high in energy) from the mitochondrial matrix to the mitochondrial inner membrane, where they release H<sup>+</sup> ions and electrons. NAD<sup>+</sup> allows the production of 3 ATP molecules and FAD the production of 2 ATP molecules upon entering the electron transport chain. Below is a description of the events that take place at the mitochondrial inner membrane, which lead to the largest yield of ATP: the electron transport chain and chemiosmosis.

#### **Electron transport chain and chemiosmosis**

Electron carriers are located on the inner mitochondrial membrane, each can receive electrons at different energy levels.

NADH<sup>+</sup>H<sup>+</sup> and FADH<sub>2</sub> are oxidized to the carriers, releasing electrons to the carriers and H<sup>+</sup> into the matrix.

Electrons "jump" from one carrier to the next, with each jump releasing some energy.

The energy released allows for the hydrogen ions at the matrix to be pumped into the small intermembrane space, against their concentration gradient.

At the ennd of the carrier chain, de-energized electrons return to the matrix and join with oxygen and hydrogen atoms, forming water as a waste product.



At the intermembrane space, hydrogen ions accumulate and the ions begin to move towards enzyme ATP synthase.

H<sup>+</sup> ions go through ATP synthase, and the enzyme becomes active, allowing for oxidative phosphorylation of ADP molecules, creating a large yield of ATP (between 36-38) molecules.



#### Figure 8.6: Electron transport chain

#### Mitochondria: site for cell respiration 8.2.5

Mitochondria are commonly referred to as the "powerhouse" of the cell. This organelle is where the Krebs cycle, electron transport chain and chemiosmosis occur, making it essential for providing the cell with the energy needed to meet its functional demands. The structure of mitochondria is very advantageous for the various functions it must accommodate. Below is an annotated diagram:



Outer membrane: separates the contents of the mitochondrion from the rest of the cell



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#### METABOLISM, CELL RESPIRATION AND PHOTOSYNTHESIS | Photosynthesis

- Inner membrane: contains the electron transport chain and ATP synthase, which carry out oxidative phosphorylation and create a large yield of energy in the form of ATP
- Intermembrane space: this small space between mitochondrial membranes allows for the rapid accumulation of protons (H<sup>+</sup>) necessary for chemiosmosis to occur
- **Cristae:** tubular regions surrounded by membranes that increase surface area for oxidative phosphorylation and the electron transport chain
- Matrix: internal cytosol-like area that contains the enzymes necessary for the Krebs cycle and link reaction

# 8.3 Photosynthesis

## 8.3.1 Light-dependent reactions

**Photosynthesis** is the process used by plants to produce their own organic substances (most commonly glucose) using light energy from the Sun and simple organic substances.

Photosynthesis involves multiple reactions, which can be simply categorized into light-dependent and light-independent. Light-dependent reactions produce intermediate compounds that are later used in the light-independent reactions to produce organic "fuels" for the plant.

Light-dependent reactions take place at the thylakoid membrane, where photosynthetic pigments are concentrated in photosystems I and II.

Light from the Sun is absorbed by chlorophyll in photosystem II (PSII).

This light energy, with the aid of an enzyme is used to split water molecules into its separate components: oxygen (released as a "waste" product), hydrogen (protons) and electrons, which are transferred to PSII  $\rightarrow$  this process is called photolysis.

Electrons in PSII are excited and "jump" up to a higher energy level, where they are received by an electron acceptor  $\rightarrow$  this is called photoactivation of PSII.

These electrons go down the electron transport chain and release energy needed to pump protoms from the stroma into the thylakoid space.

Proton accumulation allows chemiosmosis to take place, leading to the activation of ATP synthase and the production of ATP. Because ADP phosphorylation depends on the use of light (photons), this process is referred to as non-cyclic photophosphorylation.


De-energized electrons are taken up by photosystem I (PSI), and are once again excited due to the absorption of light and take up by an electron acceptor. From here, two things may occur:

- Electrons enter the electron transport chain once more, to promote further yield of ATP in a process referred to as cyclic photophosphorylation.
- If ATP levels are high enough, electrons are taken up (reduced) electron carrier NADP to form NADPH, and carried away to the stroma to be used in the light-independent reactions.

Overall, the light-dependent reactions produce:

- ATP
- NADPH
- O<sub>2</sub> (released as a by-product)

Figure 8.8



## 8.3.2 Light-independent reactions

Below is an explanation of the light-independent reactions, which take place in the chloroplast's stroma and are commonly known as the Calvin cycle, producing glucose and other organic compounds:

Products made in the light-dependent reactions (ATP and NADPH), as well as carbon dioxide molecules that diffuse into the chloroplast move to the stroma.

The cycle begins and ends with 3 5-carbon molecules called ribulose bisphosphate (RuBP).



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#### METABOLISM, CELL RESPIRATION AND PHOTOSYNTHESIS | Photosynthesis

RuBP undergoes carbon fixation with the aid of enzyme rubisco, which catalyzes the molecules' carboxylation using three carbon dioxide molecules. This yields 3 6-carbon intermediate molecules.

These intermediate molecules are so unstable that they immediately split into 6 3-carbon molecules called glycerate-3-phosphate (G3P).

G3P is reduced by NADPH and phosphorylated bu ATP, yielding 6 3-carbon triose phosphate (TP) molecules.

One TP molecule leaves the cycle and can be synthetisized into glucose and other organic compounds as the cycle is repeated and TP accumulates.

The remaining five TP molecules are phospholurated by ATP adn rearranged back into the intial 3 RuBP molecules the cycle started with



Figure 8.9: Calvin Cycle



## 8.3.3 Chloroplasts: the site for photosynthesis

Chloroplasts, organelles that are unique to plant cells and eukaryotic algae, are the site where photosynthesis reactions take place. Their structure clearly adapts to their function as can be seen in the annotated diagram below:

Figure 8.10: Chloroplasts Outer membrane Inner membrane Stroma (with 70s ribosomes) Granum Thylakoid Lamella

Skill: Annotation of a diagram of a chloroplast to indicate the adaptations to its function.

- Thylakoids: the small, disc-shaped structures increase the surface area for light absorption by photosystems I and II
- Thylakoid space: the small space inside thylakoids allows for more rapid accumulation of protons to create a concentration gradient for chemiosmosis
- Stroma: cytosol-like substance where all the enzymes needed for the Calvin cycle are found
- **Double membrane:** isolates the working parts of enzymes within the chloroplasts from the surrounding cytosol
- Lamella: connect granules and further increase surface area for light absorption





# **PLANT BIOLOGY**



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# 9.1 Transport in xylem

**Transpiration** is the loss of water, in for water vapour, from the leaves and stems of plants.





- Palisade mesophyll is the main photosynthetic tissue, and makes up the top part of the leaf
- **Spongy mesophyll** is the less densely packed mesophyll that provides the surface of gas exchange. It faces the bottom side of the leaf
- Waxy cuticle prevents exessive water loss from both the top and bottom side of the leaf
- Epidermis (both upper and lower), produces the waxy cuticle and prevents water loss
- Stoma are small gaps in the lower epidermis that allow gas exchange and therefore minimize water loss

In general, the inside of the leaf needs to be as moist as possible to allow good gas diffusion from and to the palisade mesophyll. Even though the leaf is designed to reduce water loss as much as possible, the stoma allow for escape of some water vapour from the plant leaves.

Xylem and phloem are two vessel structures in a plant that ensure that allparts of the plant are supplied with water and nutrients.



**Xylem** is the main water transport vessel and can be recognized in the picture by its large hollow spaces.

**Phloem** is the main vessel for transport of organic molecules, such as sugars and amino acids.

**Cambium** makes up the interface of xylem and phloem and can become either one of them.

## 9.1.1 Water transport

#### Root

The main part of water absorption occurs in the root of the plant. The soil contains a lot of water, but also mineral ions.

With the large surface area of root, the plant is able to absorb a lot of water from the soil by osmosis, as the root cells are more saturated in sugars and ions compared to the soil.

The mineral ions can only be absorbed through active transport since the concentration of ions is greater in the root than in the soil (think about the volume of a root compared to the volume of the total soil in a forest - which is more easily saturated?).

There are mitochondria and protein pumps in the root cells which allows the plant to take up sodium, potassium and other minerals through active transport.

#### Stem

Xylem is the water transporting vessel in the plant.

It is a tubular structure, made of dead plant cells that have fused together.

The primary xylem originates in the root and is strengthened by thick cell wall and lingin.

The rest of the xylem is also often impregnated with lingin (these plants seem woody) which helps the xylem cells resist the inward collapse produced by very low pressures.

Water is pulled to the top of the plant due to water loss from the leaves.



### PLANT BIOLOGY | Transport in xylem

Recall that the cohesive and adhesive properties of water allow it to make a continuous stream and adhere to the walls of xylem (respectively).





Xylem cells are dead which allows movement of water. Lumen is filled with sap. The ring shaped structures are thickenings of cell wall surrounded by lingin. Pores in the outer cell wall allow for water to move in and out of the xylem to the surrounding tissues.

There are three main assumptions about water transport that can be tested by simple experiments:

- 1. Water has adhesive properties
- 2. The cell wall contains small capillaries through which the water moves
- 3. Evaporation of water leads to tension in the xylem

Experiments that test these three asumptions:

- 1. If a glass capillary (hollow) is placed in a bowl of water, the water will slowly be pulled up the capillary. This is not the case if you replace water by mercury which doesn't have adhesive properties.
- 2. If you dip a strip of paper in water (and paper is made of cellulose), the water will slowly move up the strip and you will see more and more of the paper getting wet
- 3. If you take a pot with many small holes, seal it and put a small tube with water excess through it, you will see the water gradually rising through the tube, because the water vapour in the pot will slowly evaporate through the holes and create tension inside the pot



# **9.1.2 Adaptations of plants to different environments**

## Saline soil

In areas with dry climates, water evaporates from the soil easily and the soil is left saturated with mineral ions.

Due to high solute concentration in such soil, the water would not be able to move through osmosis into most plants.

Halophytes are plants adapted to saline soils as they keep the solute concentrations inside their roots, higher than the concentration of the soil.

They do this by keeping high concentration of potassium an sugars in their cytoplasm (not sodium since high sodium impairs metabolic functions).

In the vacuoles, they can keep high levels of Na<sup>+</sup> since there are no metabolic precesses going on there.

In this way, halophytes maintain high ion concentrations so that the little water from the soil would still enter the plant by osmosis.

### Desert

Xerophytes are plants adapted to life in dry habitats. Cacti are examples of xerophytes and they thrive in deserts.

There are several adaptations that will help them achieve this:

- They have a thick waxy cuticle that prevents excessive water loss.
- Their stems are very upright, so that the largest parts of the stems are only exposed to sunlight in the morning and evening (when the sun is not straight above them).
- Their stomata have the so called CAM physiology, which allows them to close during daylight and open at night, which is the opposite of what happens in normal plants (recall that stomata are needed for gas exchange during photosynthesis that occurs during daylight).
- The areas of their leaves is reduced, so cacti have spines instead of true leaves.

An example of such a plant is marram grass.





The plant is tightly attached to a long capillary tube that has a water source.

There is an air bubble inside the capillary that indicates how much water the plant has taken up.

As the water transpires off of the plant's leaves, the water is taken up into the capillary and the bubble moves.

In order to calculate a precise amount of water that was taken up, the measurements should be repeated several times.

# 9.1.3 Factors affecting transpiration rate

### Temperature

At higher temperatures, the water molecules evaporate more easily from the surface of the leaves.

At higher temperatures, air molecules around the leaf move faster, so the relative humidity is also lower.



#### PLANT BIOLOGY | Transport in xylem

At too high temperatures, the leaves' stomata may close, and the transpiration decreases slightly.

How to measure this factor using a potometer?

Using a heat lamp directed at the plant, and a thermometer to measure leaf temperature, you can vary the temperature of the plant

## **Humidity**

Humidity refers to the amount of water molecules in the air.

The higher the humidity, the smaller the concentration gradient between the moist mesophyll and humid environment, so the transpiration decreases.

How to measure this factor using a potometer?

- A plastic bag around the plant will make sure no air escapes
- By spraying moisturising spray in the bag, or by adding silica bags to the plastic bag, you can increase/decrease the humidity around the plant
- The relative humidity can be measure using a hygrometer

### Wind

If the air around the leaf is still, the water vapour in the air will not move far away from the leaf and the humidity will increase.

The higher the wind speed, the lower the humidity around the leaf, and the higher the transpiration.

At too high wind speeds, the stomata may close.

#### How to measure this factor using a potometer?

- An electric fan can produce movement of air around the plant, and you can vary its velocity by moving the fan closer or further away from the plant
- An anemometer is used to measure the exact speed of air movement



# 9.2 Transport in phloem

**Phloem** is the vessel transporting organic compounds within the plant, and this term is often substituted by the term phloem sieve tube.

## 9.2.1 Structure

Phloem is made of live cells which break down their nucleus and most of the organelles.

The walls in between the cells contain many holes called the sieve plate, which both help maintain the structure of the tube, and allow for sap to pass through either direction.

The membrane of cells contains many protein pumps that load and unload sucrose.

The membrane also contains the so called plasomodesmata, which are tight connections between the phloem cells and the companion cells in the vicinity.

## 9.2.2 Function

Phloem brings food molecules to all parts of the plant.

The photosynthetic parts of the plant produce food molecules and are therefore called sources.

In the sources, sucrose is being loaded into the phloem through active transport.

Storage parts of the plants, such as roots (carrots), tubers (potatoes) or fruits, are also called sinks, as this is where the sucrose gets unloaded from the phloem.

## 9.2.3 Hydrostatic pressure

Hydrostatic pressure is the pressure within the liquid and is maintained by different solute concentrations in the liquid and surrounding spaces.

In a plant, sources are rich in sugars leading to uptake of water from the cells surrounding the source. This leads to an increase in hydrostatic pressure.

At the sources, the solute concentrations are low, so the water can leave the phloem and enter the surrouinding tissues.



This leads to a lower hydrostatic pressure and therefore a pressure gradient between sieve tubes at sources and sinks.

## 9.2.4 Co-transporter

Sucrose I loaded from the source into the phloem via active transport.

This active transport occurs with the help of co-transporter proteins which use the gradient of ions to move molecules like sucrose from the tissue to phloem.

One protein pump, pumps hydrogen ions  $(H^+)$  to the tissue containing high levels of sucrose, while the co-transporter makes use of this ion gradient to move the sucrose into the phloem (recall that ATP synthase also uses the energy of the proton flow to make ATP).

Companion cells can help with this process as they take up the sucrose from the source, and then pass it onto the sieve tube.

# 9.2.5 Measuring the rate of phloem transport

Aphids are small animals that feed on the sap of plants.

They do this by piercing the plant with their stylets until they reach the phloem sieve tube.

If the scientist supply the plant with carbon dioxide containing radioactive carbon, the plant makes radioactive sucrose which can be detected.

Once the aphid pierces the phloem, the stylet is cut off, and the sucrose that comes out can be collected.

The time taken for the radioactive sucrose to move to different parts of the phloem can be used as a measure of phloem transport rate.



# 9.3 Reproduction of flowering plants



Figure 9.5: Seed



Seed contains the embryo of a plant and supplies the food and water needed for its development. The seed is made of modified leaves called cotyledons.





Each of these three processes needs to occur for the plant to reproduce successfully!

# 9.3.1 Flowering and day length

Different plants flower at different times of the year.

Short day plants flower in winter, while ling day plants flower in summer.

It is the length of the dark period that determines when the plant will flower.

Plants can be stimulated to flower "out of their season" by varying the amount of time when they are exposed to light vs dark.

# 9.3.2 Mutualism and pollination

Most of the flowering plants depend on insects and other animals for reproductions.

This is called a mutualistic relationship, since the animal that pollinates the flower feeds on the nectar from the plant, and in return, it transfers pollen from one plant to another (or within one plant).

Often, one specific species of insect pollinates one specific plant.

An example of this is a species of bee that feeds on vanilla orchid nectar and therefore pollinates only vanilla orchids by crossing from one plant to another in search for food.



# 9.4 Growth in plants

- **Meristems** are regions of plants containing undifferentiated cells which continuously divide and grow. Meristems at the tip of the root and shoot are called apical meristems.
- **Determinate growth** can be observed in animal species, where the embryo does not grow indefinitely, but has a determined number of legs, arms, organs, etc, to grow.

**Indeterminate growth** is seen in plants, where the apical meristem can continually provide new cells for further growth of the plant.

# 9.4.1 Growth of the shoot

Shoot of the plant is defined as the stem with leaves. As mentone earlier, the shoot contains the cells of apical meristem which continuously divide

During these divisions, some of the cells get displaced to the sides of the meristem and therefore stop dividing.

Instead, they grow and differentiate in order to produce stem and leaf tissues.

Leaves are produced when the meristem cells get displaced to the side of the apical meristem and form bumps.

These bumps, called leaf primordia, are differentiated cells that continue to divide and grow until they form full grown leaves.

Auxin is a plant hormone that controls growth in the shoot tip.

**Phototropism** is directional growth, guided by the brightest source of light.



## 9.4.2 Auxin and plant growth

There are photosensitive pigments in plant cells, called photopropins that differences in light intensity.

Once activated, the photopigments also activate proton pumps which pump  $H^+$  ions into the cell wall.

In the cytoplasm, auxin is a negatively charged molecule.

The positive charge of the wall attracts auxin molecules to move towards the wall, where they bind a hydrogen ion and then diffuse into the next cell.

Once in a cytoplasm again, auxin loses the H<sup>+</sup> ion and becomes negatively charged.

It can then again move to the next cell, and therefore redistribute to the shadier side of the plant.

Once in the shadier parts of the plant, auxin binds its receptor and activates genes responsible for secretion of more  $H^+$  ions into the cellulous cell wall.

With the extra  $H^+$  ions in the cell wall, the acidity increases, and the cellulose loosens up, allowing the plant to elongate, which leads to the general movement of the plant towards the light.



Phalaris canariensis: cotyledons after exposure in a box open on one side in front of a south-west window during 8 h. Curvature towards the light accurately traced. The short horizontal lines show the level of the ground.





## 9.4.3 Germination

In order for the embryo shoot to emerge from the seed (recall section 9.3), the seed needs to be exposed to optimal conditions

Three factors play a major role:

Water to rehydrate the plant

Oxygen to start up cell respiration

Warmth to provide optimal temperature for enzyme activity

You can design an experiment where you provide your seed with enough water, oxygen and heat. In order to determine the effect of each factor, you should keep the other two constant, and vary the third. Here is an example.

#### Effect of water

Example,

Take 2 sealed flasks and place them at room temperature.

Make sure that the flasks are large enough to provide enough oxygen for several days when sealed.

Before sealing, add a moist cotton wool at the bottom of one, and a dry cotton wool at the bottom of the other.

Place the seeds on the cotton wool, and seal the flasks.

Examine the germination over a period of a few days.

## 9.4.4 Micropropagation

**Micropropagation** is a term used to describe propagation of plants (growth) from a single small piece of plant tissue.

First, a small piece of plant tissue is taken from the shoot tip of a plant.

The piece of tissue is sterilised and placed in a sealed flask containing nutrient rich agar get with abundant auxin.



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The plant is allowed to grow into a shapeless lump of tissue called callus which can be split for extended growth.

A piece of callus can be transferred to auxin poor agar, containing other hormones needed for shoot and root development.

Once the plantlet develops, it can be transferred to soil to continue its growth into a proper plant.

## Advantages of micropropagation

- It speeds up the process of plant propagation by using only small pieces of tissue and supplying them with appropriate hormones
- The shoot tips usually are virus free, even though the rest of the plant can be infected, so virus-free strains can be produced
- The costs of plant production is decreased, and plants needn't be taken out of their natural habitat



PLANT BIOLOGY | Growth in plants

