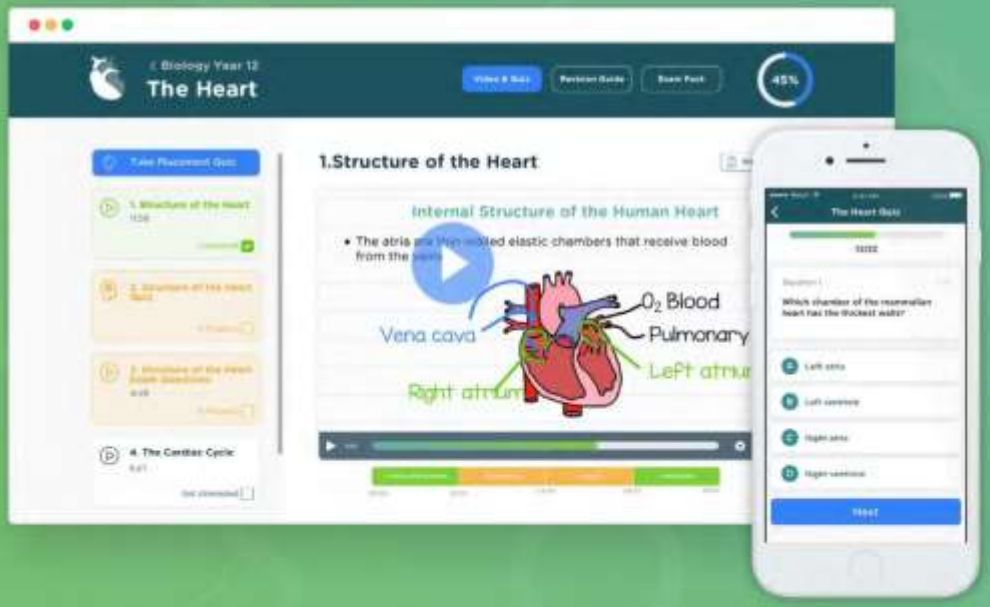
SNAPREVISE

A-LEVEL AQA BIOLOGY SUMMARY SHEETS

SNAPREVISE



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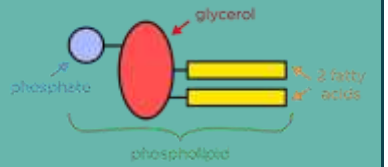
Lipids

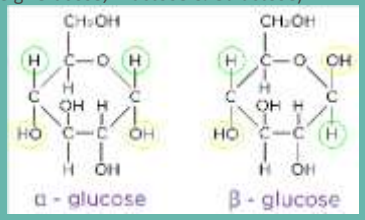
- Fatty acids can be:
  - Saturated – there are no C=C bonds and the molecule has as many hydrogen atoms as possible.
  - Unsaturated – there is at least one C=C bond, therefore the molecule contains fewer hydrogen atoms than is maximally possible.
- A triglyceride molecule is formed by joining one molecule of glycerol to three fatty acids through three condensation reactions, forming ester bonds.
- Triglycerides have key roles in respiration and energy storage due to its insolubility and high carbon to hydrogen ratio.

- Phospholipids replace one of the fatty acid chains in triglycerides with a phosphate molecule.
- The hydrophobic tails and hydrophilic heads of phospholipids allow them to form phospholipid bilayers.



Carbohydrates

- Monosaccharides are the simplest carbohydrates, consisting of only one sugar molecule (e.g. Glucose, Fructose & Galactose).



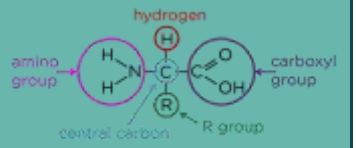
- Glucose is a hexose sugar with 2 isomers
- Disaccharides are sugars that are composed of two monosaccharides joined together in a condensation reaction, forming a glycosidic bond.

Disaccharide	Constituent monosaccharides
Maltose	× **α-glucose**
Sucrose	**α-glucose** and **fructose**
Lactose	**β-glucose** and **galactose**

- Polysaccharides are formed by many monosaccharides joined together.
  - Amylose, amylopectin (starch) is the main polysaccharide energy store in plants, is composed of α-glucose.
  - In animals, the polysaccharide energy store is called glycogen, composed of α-glucose.
  - Cellulose is a structural component of plant cell walls, composed of long unbranched chains of β-glucose.

Proteins

- Amino acids are the monomer units used to make proteins.
- The 20 naturally occurring amino acids only differ in their R groups.
- Dipeptides are formed when two amino acids are joined together by a condensation reaction, forming a peptide bond.
- A polypeptide is a polymer made of many amino acids joined together by peptide bonds.

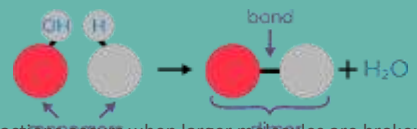


- A protein may contain one or more polypeptide chains.
- There are four structural levels:

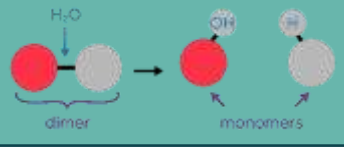
Level	Definition	Bond type
Primary	The specific sequence of amino acids in a polypeptide chain	Peptide bonds
Secondary	The curling or folding of the polypeptide chain into **α**-helices and **β**-pleated sheets due to the formation of hydrogen bonds	Hydrogen bonds
Tertiary	The overall specific 3-D shape of a protein, which is determined by interactions between R groups and the properties of R groups	Hydrogen bonds Ionic bonds Disulphide bridges

Monomers & Polymers

- Monomers are individual molecules that make up a polymer.
- Polymers are long chains that are composed of many individual monomers that have been bonded together in a repeating pattern.
- Condensation Reactions occurs when two molecules combine to form a more complex molecule with the removal of water.



- Hydrolysis Reactions occurs when larger molecules are broken down into smaller molecules with the addition of water.



Quaternary	The specific 3-D shape of a protein that is determined by the multiple polypeptide chains and/or prosthetic groups bonded together	Hydrogen bonds Ionic bonds Disulphide bridges

Biochemical Tests

Molecule	Reagent	Positive result
Reducing sugars	Benedict's reagent → Heat	Red/orange precipitate
Starch	Iodine in potassium iodide solution	Blue/black
Non-reducing sugars	Hydrochloric acid → Heat Sodium hydrogencarbonate Benedict's reagent → Heat	Red/orange precipitate
Proteins	Sodium hydroxide Copper (II) sulphate	Purple
Lipids	Ethanol Water → Shake	Cloudy white

Enzymes

- Enzymes are biological catalysts that speeds up the rate of reaction and remains unchanged and reusable at the end of the reaction.
- They lower the activation energy of the reaction.
- The lock and key model proposed that each substrate is a key that only fits a specific lock or enzyme. The alternative induced fit model has been proposed (below)

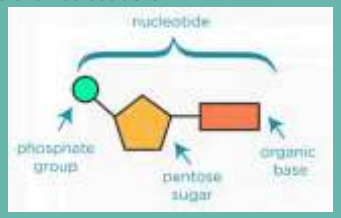
- The specificity of enzymes is due to the tertiary structure of its active site, allowing complementary binding to substrates.
- Enzymes catalyse both intracellular and extracellular reactions that determine structures and functions from cellular to whole organism level.
- Factors affecting enzyme activity include:

pH: Temperature Enzyme concentration

Substrate Competitive & non-competitive inhibitor concentration concentration

DNA & RNA

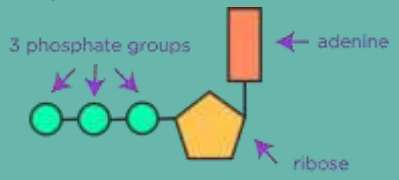
- DNA & RNA are both polynucleotides.
- The basic structure of a nucleotide is:



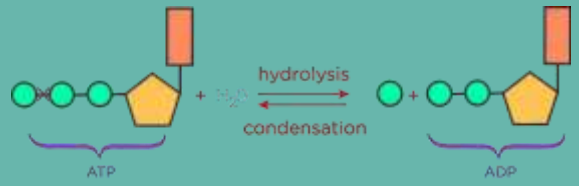
	DNA	RNA
Number of Strands	Two antiparallel strands	One strand
Length	Very long	Relatively short
Pentose Sugar	Deoxyribose	Ribose
Nitrogenous Bases	Adenine, Cytosine, Guanine & Thymine	Adenine, Cytosine, Guanine & Uracil
Function	Store genetic information	Transfer genetic information & forms ribosomes with proteins

ATP

- The structure of ATP is:



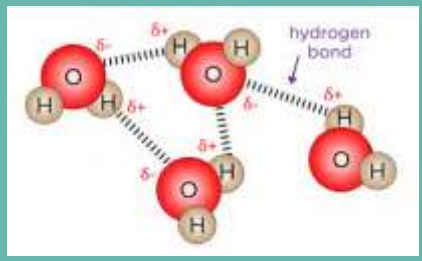
	ATP → ADP	ADP → ATP
Reaction type	Hydrolysis	Condensation
Enzyme involved	ATP hydrolase	ATP synthase
Energy profile of reaction	Releases energy	Requires energy



- The hydrolysis of ATP can be coupled to energy-requiring reaction and used to phosphorylate compounds.
- The condensation of ADP to form ATP can occur during respiration and photosynthesis.

Water

- Water molecules consist of 2 hydrogen molecules covalently to an oxygen molecule.

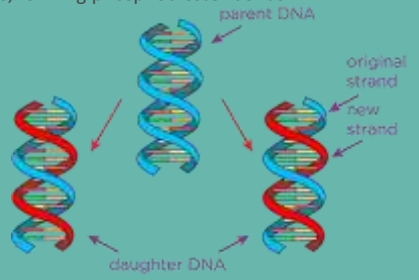


- The molecules are slightly polar because the oxygen nucleus pulls the shared electrons away from the hydrogen nuclei. Giving the oxygen nuclei a δ- charge, and the hydrogen nuclei a δ+ charge.
- The polarity of water causes attraction between water molecules. This force of attraction is called a hydrogen bond.

Property of water	Why it is useful
Liquid medium	Provides habitats for aquatic organisms, medium for chemical reactions & used for transport
Important metabolite	Used in hydrolysis & condensation reactions
High specific heat capacity	Keeps aquatic & cellular environments stable

DNA Double Helix & Replication

- Polynucleotides are polymers made up of many nucleotide monomers joined together by a series of condensation reactions, forming phosphodiester bonds.
- The DNA double helix is held together by hydrogen (H) bonds between complementary base pairs.
  - 2 H bonds between Adenine & Thymine
  - 3 H bonds between Cysteine and Guanine
- Semi conservative replication is the method in which DNA replicates, creating two molecules of DNA that consist of one original DNA strand and one newly synthesised DNA strand.
  - DNA helicase breaks H bonds between the two strands
  - Free nucleotides complementary base pair to the exposed strands
  - DNA polymerase catalyses condensation reactions to join adjacent nucleotides, forming phosphodiester bonds.



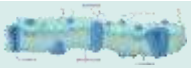
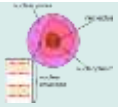
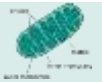
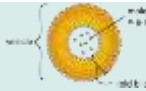
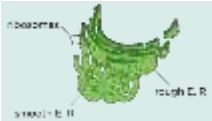
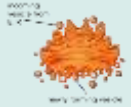
Inorganic Ions

- Inorganic ions are atoms or molecules with an electric charge, containing no carbon.
- Cations are positively charged ions
- Anions are negatively charged ions
- Inorganic ions occur in solution in the cytoplasm and body fluids of organisms, some in high concentrations and others in very low concentrations
- Each type of ion has a specific role, depending on its properties
  - Hydrogen ions determine the pH of bodily fluids. The higher the concentration, the lower the pH
  - Iron ions are essential components of the prosthetic group in haemoglobin and bind to oxygen
  - Sodium ions are used in the co-transport of glucose and amino acids across cell membranes
  - Phosphate ions are essential components of DNA, RNA & ATP

High latent heat of vaporisation	Evaporation has a cooling effect on organisms
Cohesion of molecules	Water is drawn up the xylem
Surface tension	Allows pond-skaters to walk on the surface
Good solvent and transport medium	Dissolves ionic and polar molecules, allowing them to easily be transported
Good reaction medium	The cytoplasm in cells is an aqueous solution where many chemical reactions happen
Incompressible	Can prevent plants from wilting & act as a hydrostatic skeleton for invertebrates

Eukaryotic Cells

- Eukaryotes include animal, plant & fungal cells.
- The following organelles are presents in eukaryotic cells:

Organelle	Structure	Function
Cell surface membrane		• Controls passage of entry of substance into the cell • Site of cell communication via receptors
Nucleus		• Stores DNA • Nuclear pores allow mRNA & ribosomes to pass through
Mitochondria		• Carry out aerobic respiration to produce ATP
Lysosomes		• Contains digestive enzymes to break down pathogens, old organelles, cells & food molecules
Ribosomes		• Site of protein synthesis
Rough endoplasmic reticulum		• Provide a large surface area for protein synthesis
Smooth endoplasmic reticulum		• Synthesise, store and transport lipids and carbohydrates.
Golgi Apparatus		• Modifies proteins • Sort, package, and transport molecules around the cell

- There are additional organelles in plants, algae & fungi:

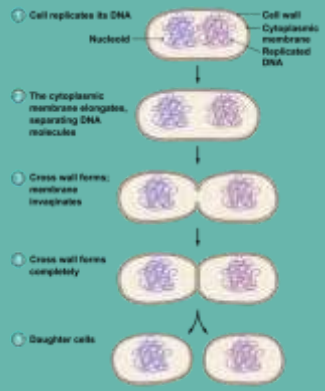
Organelle	Structure	Function	Present in which organism

Prokaryotic Cells

- Prokaryotes are smaller and simpler than eukaryotes.

Feature	Eukaryotic Cell	Prokaryotic Cell
Nucleus	Present	Absent
DNA	Linear and packaged into chromosomes in nucleus	Circular and freely floating in cytoplasm
Cell Membrane	Present	Present
Membranebound organelles	Present	Absent
Ribosomes	Present (80S)	Present (70S)
Cell Wall	Sometimes (cellulose or chitin)	Present (peptidoglycan)
Chloroplasts	Sometimes	Absent
Flagellum	Absent	Sometimes
Capsule	Absent	Sometimes
Plasmid	Absent	Sometimes

- Bacteria replicate by binary fission.



Methods of Studying Cells

- There are 3 main types of microscopes used to observe cells:

	Light Microscope	Scanning Electron Microscope	Transmission Electron Microscope
Medium	Light Beam	Electron Beam	Electron Beam
Dimensions	2D	3D	2D
Max Magnification	X1,500	X200,000	X2,000,000
Max Resolution	200 nm	20 nm	0.1 nm

- Magnification is how much bigger the image is compared to the original object viewed with the naked eye
- Magnification = (size of image)/(size of object)
- Resolution is how well a microscope distinguishes between two points that are close together.
- Cell fractionation can be used to separate organelles.
  - Homogenisation - grinding cells release the organelles into solution
  - Filtration - separates organelles & debris
  - Ultracentrifugation - using a centrifuge the organelles are separated out in order of mass

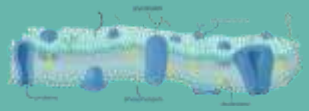
Cell Division

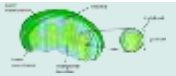
- Within multicellular organisms, not all cells retain the ability to divide
- The eukaryotic cell cycle has three main stages:
- Interphase consists of two growth phases (G_1&G_2) and a DNA synthesis stage (S). The cell may exit the cell cycle at G0
- Mitosis is the nuclear division
- Cytokinesis is when the cell splits in two, forming two identical daughter cells.

Stage	Description	

Cell Membranes

- Cell membranes act as barriers and can control what passes into and out of cells and organelles
- The cell membrane is composed of phospholipids, proteins, glycoproteins, glycolipids and cholesterol.
- Cholesterol has a hydrophilic end and a hydrophobic end & regulates membrane fluidity by intercalating between the phospholipids.

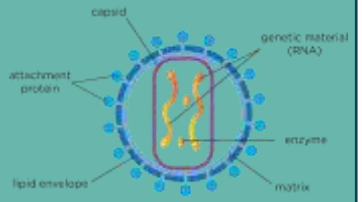


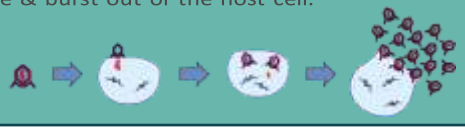
Chloroplasts		• Site of photosynthesis	Plants & algae
Cell vacuole		• Maintains cell structure • Act as a temporary energy store	Plants
Cell wall		• Provides support & mechanical strength	Plants & algae
			Fungi

- In complex multicellular organisms, eukaryotic cells become
- Specialised for specific functions.
- Specialised cells are organised into tissues, tissues into organs and organs into systems.

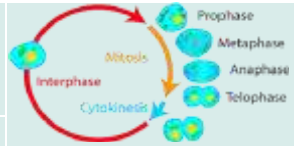
Viruses

- Viruses are acellular and non-living.
- The basic structure of viruses is:
- Viruses replicate by binding to the host cell, injecting their genetic material into the cell, using the host's machinery to replicate & burst out of the host cell.





Prophase	DNA condenses & coils, nuclear envelope breaks down, centrioles move to opposite poles
Metaphase	Spindle fibres attach to centromeres & chromosomes line at the equator
Anaphase	Centromeres divides, chromatids move to opposite poles
Telophase	Chromosomes uncoil, nuclear envelope reforms



- Cancerous cells have uncontrolled cell division and hence have a modified cell cycle – one that repeats too quickly.
- Treatments for cancer involve disrupting the cell cycle (chemotherapy) by stopping DNA synthesis or by changing the cytoskeleton in mitosis

SNAPREVISE

GAS EXCHANGE, DIGESTION & MASS TRANSPORT

SUMMARY SHEET

Passive Transport

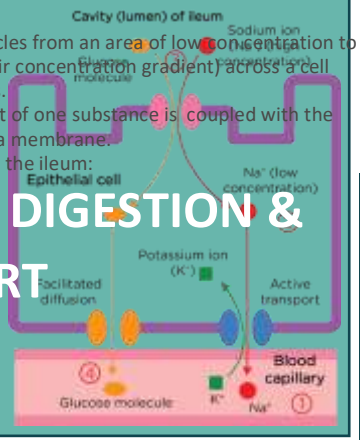
- Passive transport involves exchange of substances without requiring metabolic energy from the cell
- Diffusion is the net movement of particles from an area of higher concentration to an area of lower concentration (down their concentration gradient).
- Facilitated diffusion is the net movement of particles down their concentration gradient across a partially permeable cell membrane via carrier or channel proteins.

- Water potential is a measure of the tendency of water molecules to move from one area to another area and describes the pressure created by these water molecules; the more dilute a solution, the higher (less negative) the water potential (Ψ).
- Osmosis is the net movement of water from an area of higher water potential to an area of lower water potential across a partially permeable membrane.
- The rate of diffusion can be increased by increasing the number of channel & carrier proteins, the surface area of the cell membrane, reducing the diffusion distance and creating a steeper concentration gradient.

Active Transport

Active transport is the movement of particles from an area of low concentration to an area of high concentration (against their concentration gradient) across a cell membrane, using ATP and carrier proteins.

- Co-transport occurs when the transport of one substance is coupled with the transport of another substance across a membrane.
- Glucose & sodium are cotransported in the ileum:



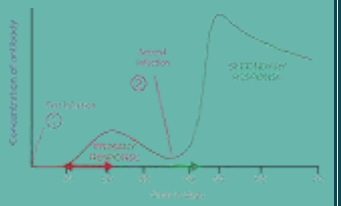
Components of the Immune System

- Antigens are any part of an organism/substance which is recognised as foreign by the immune system and goes on to trigger an immune response.

Cell	Function

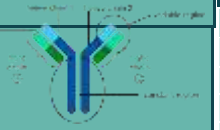
Primary & Secondary Response

- The primary immune response is when a pathogen infects the body for the first time the initial immune response is slow
- The secondary immune response is a more rapid and vigorous response caused by a second or subsequent infection by the same pathogens. This is due to the presence of memory cells.



lymphocytes in response to the presence of the corresponding antigen.

- Antibodies agglutinate pathogens by forming antigen-antibody complexes, leading to phagocytosis & neutralise toxins.



Cell-mediated Immunity

- Antigen from the pathogen is displayed on the cell surface of body cells or phagocytes after phagocytosis
- T cells with the correct specific receptor bind with the antigen and are activated
- They divide by mitosis (clonal expansion) and differentiate into T helper, cytotoxic and memory cells.

Humoral immunity

- The humoral response is best at fighting pathogens which are free in the bodily fluids
- Free antigen binds to a complementary B cell receptor, activating the B cell (clonal selection)
- The pathogen is endocytosed, and the antigen presented on the plasma membrane
- T helper cell binds to the presented antigen and stimulates the B cell to divide by mitosis (clonal expansion)
- The B cell differentiates to plasma and memory cells

Vaccination

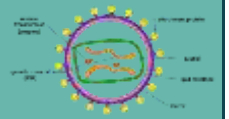
Using Monoclonal Antibodies

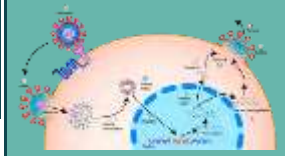
- Drugs can be attached to monoclonal antibodies, in order to ensure the delivery of the drug to specific cell types e.g. cytotoxic drug to a cancer cell
- Disease diagnosis can occur by testing for the presence of specific pathogen antibodies in the blood.
- Monoclonal antibodies are also used for pregnancy testing
- Measurement & diagnosis of antigen occur in the ELISA test where different monoclonal antibodies are bound to the surface of a well. They attach to antigen present in a sample, allowing the attachment of a detection antibody. An enzyme attached to the detection antibody digests a substrate, which is added, causing a colour change. The colour intensity corresponds to the amount of the antigen present in the sample
- Ethical considerations: treatment may cause death (risky), use of animals for production may cause harm, human trials

Immunity	Example
Natural Active	Direct contact with pathogen
Natural Passive	Antibodies through breastmilk
Artificial Active	Vaccination
Artificial Passive	Injection of antibodies

Human Immunodeficiency Virus (HIV)

- HIV replicates in T helper cells, causing the symptoms of AIDs due the to decreased cell count. The compromised immune system leads to the risk of serious infections.

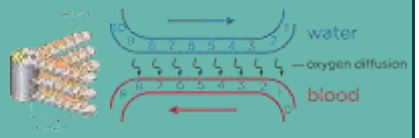




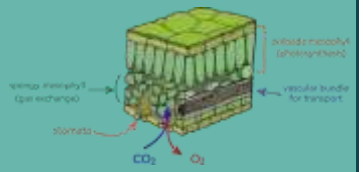
- Antibiotics kill bacteria by targeting bacteria specific enzymes or organelles. They are ineffective against viruses due to the virus using the host's machinery.

Gas Exchange

- Single celled organisms can exchange oxygen and carbon dioxide directly through their plasma membrane via diffusion.
- Insects exchange gas in their tracheal system. Air enters via spiracles, travels through trachea and tracheoles, delivering oxygen directly to every tissue.
- Gas exchange in fish occur via gills. The orientation of the gill filaments and lamellae ensures that the water flowing over them moves in the opposite direction to the flow of blood through the capillaries (countercurrent flow), maintaining a diffusion gradient.

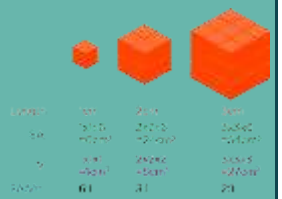


- Gas exchange in dicotyledonous plants occurs in the leaves. The stomata can open to allow gases diffuse in and out of the leaf. The mesophyll cells have a large surface area for rapid diffusion.
- Gas exchange can lead to water loss. Plants can control the opening of their stomata to limit this, and xerophytes may have additional adaptations such as: hairs, waxy cuticle, small leaves, sunken stomata, rolled leaves. Insects can also control water loss but controlling open and closing of their spiracles, hair around spiracles and a waterproof, waxy cuticle.



Surface Area to Volume Ratio

- The greater the size of an organism, the smaller its surface area: volume ratio
- Larger organisms therefore require specialised exchange surfaces and transport mechanisms to meet their metabolic requirements
- Specalised exchange surface have: a large surface area, thin barriers and associated transport systems to maintain a steep diffusion gradient.
- Also, organisms with a higher metabolic rate require more nutrients and produce more waste, therefore require a specialised exchange surface



Digestion

- During digestion, large biological molecules are hydrolysed to smaller molecules that can be absorbed across cell membranes
- Digestion enzymes in mammals includes:

Enzyme		Substrate	Product(s)
Amylase		Starch	Maltose
Membranebound disaccharidases	Maltase	Maltose	α-glucose molecules
	Sucrase	Sucrose	Glucose & fructose
	Lactase	Lactose	Glucose & galactose
Lipase		Lipids	Monoglyceride & fatty acids
Endopeptidases (pepsin, trypsin & chymotrypsin)		Hydrolyse peptide bonds in the middle region of proteins	Produce several polypeptide chains
Exopeptidases		Hydrolyse peptide bonds on terminal amino acids	Release single amino acids and dipeptides
Membrane-bound dipeptidases		Dipeptides	Single amino acids

- The ileum is the final section of the small intestine where both hydrolysis and absorption occurs.
- Bile salts made by the liver, emulsify lipids in order to increase the surface area of the lipids, for greater access to lipases.
- Micelles are the products of lipase digestion that remain in association with the bile salts to form structures. The micelles travel to the ileum where, upon contact with the surface of ileum epithelium cells, they are broken down. This releases the non-polar monoglyceride and fatty acids, which diffuse straight into the epithelial cell.
- Amino acids and carbohydrates are absorbed via cotransportation with sodium.

Human Gas Exchange System

- In humans, gas exchange occurs via the lungs
- The alveolar epithelium is adapted for gas exchange by having a large surface area, good blood supply, thin walls & elastic fibres which help recoil

- Ventilation is the process of breathing in (inspiration) and out (expiration).
- Inspiration: external intercostal muscles contract, rib cage moves up & out, diaphragm contracts, volume of the thorax is increased, atmospheric pressure is greater than pulmonary pressure and air is forced into the lungs.

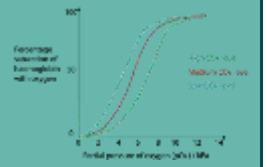
- Expiration: internal intercostal muscles contract, ribs move down and inwards, diaphragm relaxes, volume of the thorax is decreased, pulmonary pressure is greater than atmospheric pressure, air is forced out of the lungs
- Pulmonary ventilation rate is the total volume of air moved into the lungs during a minute.
- Tidal volume is the volume of air moved in and out of the lungs with a normal breath.
- Breathing rate is the number of breaths per minute.
- Pulmonary Ventilation Rate (dm^3min^{-1}) = Tidal Volume (dm^3) × Breathing Rate (min^{-1})

Mass Transport in Animals

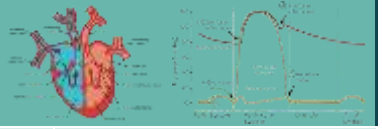
- Red blood cells transport oxygen using the protein haemoglobin

$$\text{oxygen} + \text{haemoglobin} \underset{\text{DISSOCIATION}}{\overset{\text{ASSOCIATION}}{\rightleftharpoons}} \text{oxyhaemoglobin}$$

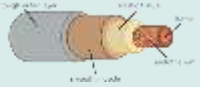
- Haemoglobin is made up of four polypeptide chains, each containing a prosthetic haem group. Each haem group binds one oxygen molecule
- Binding of the first O2 molecule causes a conformational change in the haemoglobin, making the haem groups more accessible to oxygen.



- Bohr affect - haemoglobin's oxygen binding affinity is inversely related to the concentration of carbon dioxide, causing the oxygen dissociation curve to shift
- The cardiac cycle is the sequence of events that occur within one full beat of the heart.
- Circulatory system:

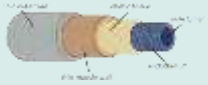


Arteries & Arterioles
transports blood away from the heart



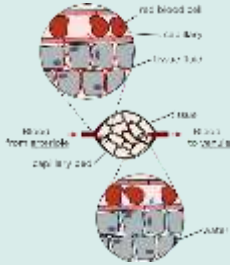
Capillaries - area of metabolic substance exchange

Veins and Venules
transports blood towards from the heart



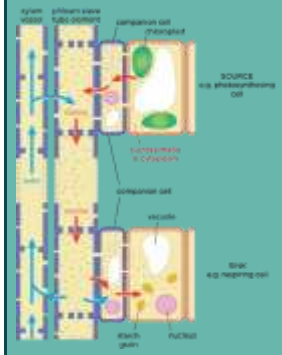
Tissue fluid formation:
Arteriole: Hydrostatic pressure > water potential
Venule: Hydrostatic pressure < water potential
Remaining fluid returns to circulation via the lymphatics system

Tissue fluid



Mass Transport in Plants

- The xylem transports water & mineral ions up the plant against gravity
- Water evaporates from the leaves creating tension (transpiration), and the cohesive nature of water moves the whole column of water up the xylem (cohesion-tension theory)
- The rate of transpiration is affected by: light, temperature, humidity & wind.
- The phloem transports assimilates from sources to sinks via translocation



- Sucrose is actively transported into the companion cells and moves via diffusion into the sieve tube followed by water. Assimilates move from area of high to low pressure (mass flow). At the sink the solutes are removed, water leaving by osmosis.
- To track the movement of sugars through the phloem, scientists' radioactive isotopes are used in tracer experiments with radioactive isotopes
- Ringing - removal of the bark and phloem, theoretically prevents translocation to the sinks below the ring

Genetic Information

- In prokaryotic cells, DNA molecules are short, circular and not associated with proteins.
- In eukaryotes, the nucleus contains very long, linear DNA molecules associated with proteins, called histones. Together a DNA molecule and its associated proteins form a chromosome.
- The mitochondria and chloroplasts of eukaryotic cells also contain DNA which, like the DNA of prokaryotes, is short, circular and not associated with protein.
- The genome is the full set of DNA found in an organism.
- The proteasome is the full range of proteins that can be synthesised from the genome.
- A gene is a section of DNA that code for polypeptides and functional RNA and are located at a fixed locus on a DNA molecule.
- A sequence of three DNA bases, called a codon, codes for a specific amino acid. The genetic code is universal, nonoverlapping and degenerate.
- In eukaryotes, sections of the nuclear DNA do not code for polypeptides (introns). Exons are sections of DNA that code for amino acid sequences.

Biodiversity

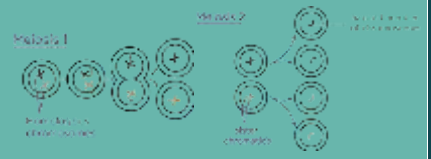
- Biodiversity is the variety of organisms in an area. It can be considered on a local or global scale.
- Species richness is a measure of the number of different species in a community.
- An index of diversity measure biodiversity taking into account species richness and the number of individuals in each species.
- Index of diversity = $(N(N-1))/(\Sigma n(n-1))$
  - N = total number of organisms of all species
  - n = total number of organisms of each species
- Farming techniques reduce biodiversity. E.g. monoculture, use of herbicide & pesticides, hedgerow removal and woodland clearance.
- Conservationists protect biodiversity with methods such as:
  giving endangered species legal protection, creating protected area & The Environmental Stewardship Scheme.
- A balance between conservation and agriculture is needed.

Causes of Genetic Variation

- Variation can arise due to mutation.
- Gene mutations are changes to the base sequence or quantity of DNA within a gene or section of DNA.

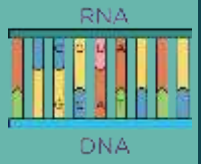
Type of gene mutation	Description
Substitution	When a nucleotide is changed to a different nucleotide. As the genetic code is degenerate, this may not change which amino acid is coded
Insertion/ Deletion	Addition/removal of one or more nucleotides into the DNA sequence. May result in a frameshift

- Mutagenic agents can increase the rate of gene mutation.
- Chromosome mutations are changes to the structure or number of whole chromosomes. E.g. failure of chromosomes to separate in meiosis (nondisjunction).
- Meiosis is also a cause of variation, as it produces 4 daughter cells that are genetically different from each other.

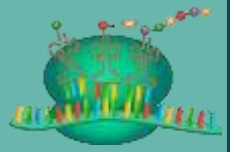


- In meiosis 1, homologous chromosomes are separated from each other, with one chromosome from each pair going into one of the two daughter cells. In the second meiotic division, the sister chromatids from each chromosome are separated.
- Variation results from independent assortment of chromosomes and crossing over during meiosis 1. Also, random fertilisation of the haploid gametes.

Protein Synthesis



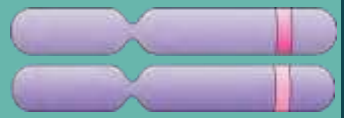
- Structure of tRNA & mRNA:
- Transcription is the process of making messenger RNA from a DNA template.
- DNA helicase breaks the hydrogen bonds between the DNA helix, free RNA nucleotides base pair with the exposed DNA template strand.
- In prokaryotes, transcription results directly in the production of mRNA from DNA.
- In eukaryotes, transcription results in the production of premRNA; this is then spliced to form mRNA.
- Translation is the process of making proteins by forming a specific sequence of amino acids based on coded instructions in mRNA. RNA polymerase catalyses phosphodiester bonds between adjacent RNA nucleotides and the mRNA strand detaches, allowing the DNA helix to reform.
- mRNA attaches to a ribosome on the rough endoplasmic reticulum, tRNA carries the corresponding amino acid to each codon on the mRNA one at a time, with an enzyme catalysing the formation of a peptide bond between amino acids using ATP, until a stop codon is reached and the peptide is released, folding into its tertiary structure.



Investigating Diversity

- Genetic diversity within or between species can be compared by looking at:
  - The frequency of measurable/observable characteristics
  - The base sequence of DNA
  - The base sequence of mRNA
  - The amino acid sequence of proteins
- Gene technology has caused a shift in methods of investigating genetic diversity from solely looking at observable characteristics
- Variation is caused by genetics & environmental factors.
- Variation can be investigated quantitatively within a species by collecting random samples (to reduce bias), calculating a mean value and the standard deviation of the data collected. Then interpreting mean values and their standard deviations.
  - Means may vary, showing variation between populations
  - A large standard deviation indicates a large amount of variation within a population

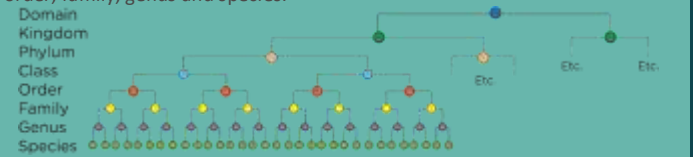
Genetic Diversity & Adaptation



- Alleles are different forms of the same gene.
- Genetic diversity is the number of different alleles of genes in a population.
- Genetic diversity is a factor enabling natural selection to occur.
- Natural selection is a mechanism of evolution by which individuals better adapted to their environment tend to survive, reproduce successfully and pass on their alleles.
- In the process of natural selection: random mutation can result in new alleles of a gene, many mutations are harmful but, in certain environments, the new allele of a gene might benefit its possessor, leading to increased reproductive success. The advantageous allele is inherited by members of the next generation. As a result, over many generations, the new allele increases in frequency in the population.
- Direction selection is a selective force that favours individuals with an extreme form of a trait and selects against phenotypes at the other extreme. E.g. antibiotic resistance. Powerful antibiotics apply a very strong selection force favouring individuals possessing resistance alleles.
- Stabilizing selection is a selective force that favours the phenotypes closest to the mean value of a trait. E.g. Human birth weight. Babies that tend to the extremes of birth weight have higher mortality rates.
- Adaptations may be anatomical, physiological or behavioural.

Classification

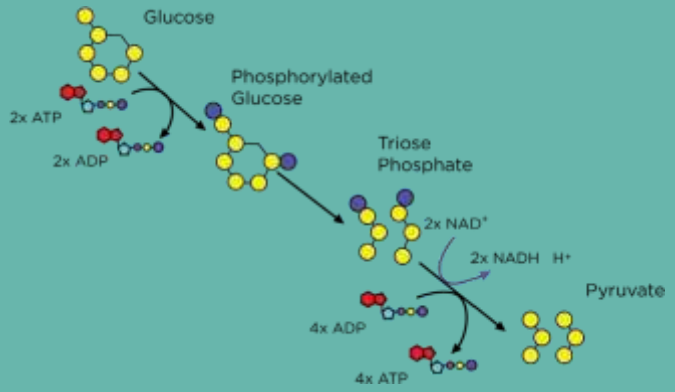
- The Biological Species Concept- a species contains all organisms that are capable of breeding together to produce living, fertile offspring.
- Courtship and mating behaviour are a vital part of species survival. Courtship behaviour enables individuals to: recognise same species members & identify mate capable of breeding.
- Classification is the process of sorting living things into groups.
- Classification hierarchy comprises the taxa: domain, kingdom, phylum, class, order, family, genus and species.



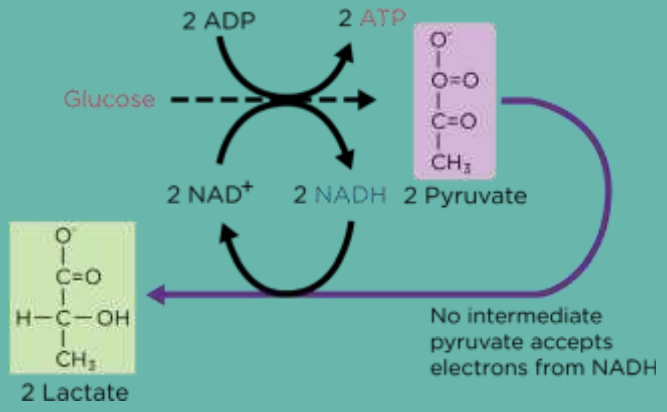
- Classifications are constantly updated as new methods are discovered to infer relationships e.g. DNA sequencing, amino acid sequencing or immunological comparisons.
- The binomial naming system names species by their genus and species name.
- Phylogeny is the study of evolutionary relationships between organisms.
- In a phylogenetic diagram, branch tips represent species at the end of their specific lineage, branching points represent common ancestors & The closer the branches, the closer the evolutionary relationship.

Anaerobic Respiration

- Respiration is the process, which occurs in living cells, that releases energy stored in organic molecules such as glucose.
- The energy released during respiration is used to synthesise molecules of ATP, which can be used as an immediate source of energy.
- The first stage of respiration is glycolysis which occurs in the cytoplasm of cells.
  - There is a net yield of 2 pyruvate, 2 reduced NAD and 2 ATP molecules



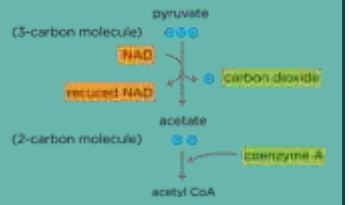
- If oxygen is not available as the final electron acceptor, glycolysis can continue in anaerobic respiration.
- Glycolysis can continue if reduce NAD is reoxidised so that NAD is available to accept a hydrogen atom again.
- In mammals, the lactate fermentation pathway is used:



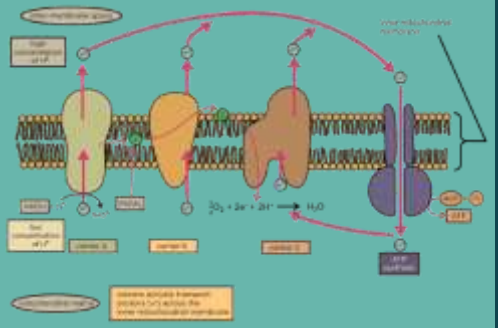
Lactate can be converted to glycogen in the liver or oxidised further to release energy, when oxygen is available.

- In plants and fungi, the ethanol fermentation pathway is used:
  - pyruvate + reduced NAD → ethanol + carbon dioxide + oxidised NAD

Aerobic Respiration



- If respiration is aerobic, pyruvate enters the mitochondrial matrix by active transport.
- Next, the link reaction occurs:
- Following the link reaction, the Krebs cycle occurs.
- The final stage of aerobic respiration is oxidative phosphorylation.
- Reduced NAD and FAD donate electrons to the electron transfer chain in the inner mitochondrial membrane. The release of energy as the electrons pass down the electron transfer chain is used to create a proton gradient across the inner mitochondrial membrane into the intermembranal space. The proton gradient is used to synthesis ATP by oxidative phosphorylation, catalysed by ATP synthase (chemiosmotic theory).



- Oxygen combines with the protons that have diffused through the ATP synthase channel and the electrons that have been passed along the electron transfer chain, acting as the final electron acceptor. It helps maintain the proton gradient for the electron transfer chain to continue.

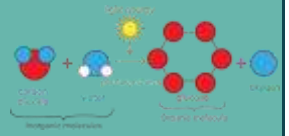
$$\tfrac{1}{2}O_2 + 2e^- + 2H^+ \rightarrow H_2O$$

Aerobic respiration produces 32 ATP. 30 more than anaerobic respiration.

- Sugars such as glucose are not the only substances that can be used as a respiratory substrate.
- Lipids release more energy than carbohydrates, due to more carbon-hydrogen bonds

Substrate	Process in respiration

Photosynthesis



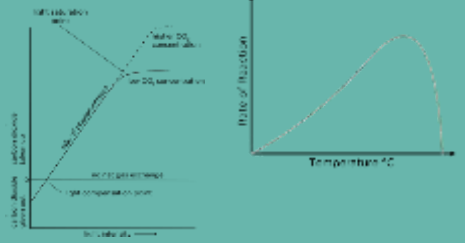
- Photosynthesis is the process in plants, from which energy from sunlight is used to convert inorganic molecules into organic molecules.
- The light-dependent reaction occurs in the thylakoids of the grana in chloroplasts
  - Photolysis of water requires light energy to break the bonds between oxygen and hydrogen atoms

$$2H_2O \rightarrow 4H^+ + 4e^- + O_2$$

Chlorophyll molecules absorb light energy via photosystem II, exciting a pair of electrons to a higher energy level, leaving the chlorophyll molecules ionized. The electron passes through an electron transfer chain to produce ATP, and reaches photosystem I.

  - The electrons replace the electrons lost in photosystem I when it absorbs light to reduce NADP with the protons created from photolysis
  - The photoionized chlorophylls electrons in photosystem II are replaced by the electrons from photolysis of water
  - Cyclic photophosphorylation only uses photosystem I, where the electrons are passed back to photosystem I rather than NADP via electron carriers, producing small amounts of ATP
- The light-independent reaction occurs in the stroma of chloroplasts
  - The Calvin cycle depends on the products from the light dependant stage
  - The fixation of carbon dioxide is catalysed by RuBisCo
  - 5 out of every 6 TP molecules are used to regenerate RuBP instead of producing hexose sugars

- The rate of photosynthesis is limited by temperature and the availability of carbon dioxide, water & light energy.
- The law of limiting factors states that at any given moment, the rate of a physiological process is limited by the factor that is at its least favourable value.



- Chromatography can be used to separate out photosynthetic pigments, identifying them by their R_f value

Lipid	Hydrolysed to fatty acids and glycerol. Glycerol is phosphorylated and converted to triose phosphate, which enters the glycolysis pathway The fatty acid part is broken down into 2-carbon fragments which are subsequently converted into acetyl CoA, also generating reduce NAD & FAD
Protein	Protein is hydrolysed to amino acids. In the liver, the amino group is removed (deamination), and the amino group is converted to urea and removed in the urine. The remaining amino acid can then be converted to an intermediate

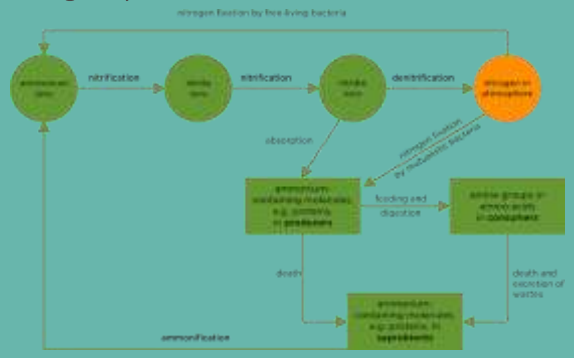
$$R_f\ value = \frac{Distance\ travelled\ by\ spot}{Distance\ travelled\ by\ solvent}$$

Biomass

- Plants synthesise organic compounds from atmospheric, or aquatic, carbon dioxide.
- Most of the sugars synthesised by plants are used as respiratory substrates. The rest are used to make other groups of biological molecules, forming the biomass of the plant.
- Biomass is the total mass of living material in a specific area at a given time
- Dry biomass shows the chemical energy store in an organism and can be measured by the process of calorimetry. A dry sample is weighed and burnt in pure oxygen within a sealed chamber, the temperature increase of the fixed volume of water is used to calculate the energy released.

Nutrient Cycles

- There is a finite supply of nutrients on Earth, which are recycled within natural ecosystems.
- The Nitrogen cycle:



- The Phosphorus cycle:

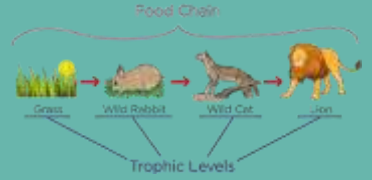
The Use of Fertilisers

- Fertilisers can be used to provide plants with minerals, particularly nitrates, to support their growth
- In agriculture systems, the harvesting of crops prevents the reintroduction of minerals to the soil
- Natural ferilisers consist of dead and decaying remains of plants, animals and their waste
- Artificial fertilisers are mined from rocks before being converted into different forms with their composition tailored for specific crops.

Effect of using fertilisers	Description
Reduced species diversity	Nitrogen-rich soils favour rapidly growing species
Leaching (pollutes waterways)	Rainwater dissolves soluble nutrients (e.g. nitrates) and carries them deep into the soil and into waterways such as streams, rivers and lakes.

Production & Productivity

- Gross primary production (GPP) is the total quantity of chemical energy stored in plant biomass, in a given area or volume.
- Net primary production (NPP) is the chemical energy store in plant biomass after respiratory losses to the environment have been taken into account
  - NPP = GPP – R
  - Where R represents respiratory loses to the environment
  - NPP, GPP & R use units of (kJ m^{-2} yr^{-1})
- The NPP is available for plant growth and reproduction. It is also available for consumers in the food chain such as herbivores and decomposers.



- Net production (N) is the total chemical energy consumers store after energy losses to faeces, urine and respiration have been taken away from the chemical energy store of the ingested plant food
  - N = I - (F + R)
  - Where N is net production, I represents the total chemical energy store in ingested food, F is the energy lost in faeces and urine, and R is energy lost to respiration. All use units (kJ m^{-2} yr^{-1})
- Primary and secondary productivity is the rate of primary or secondary production, respectively. It is measured as biomass in a given area in a given time e.g. kJ ha^{-1} $year^{-1}$
- The percentage efficiency of energy transfer from one tropic level to another can be calculated as

$$\left(\frac{\textit{energy available after the transfer}}{\textit{energy available before the transfer}}\right) \times 100$$

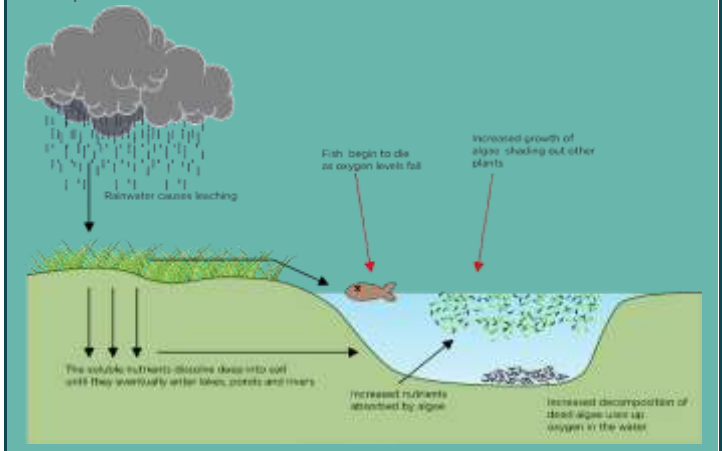
- Farming practices increase the efficiency of energy transfer to increase yields by:
  - Reducing respiratory loses in a human food chain e.g. reduce movement of animals
  - Simplifying food chains to reduce energy loss to non-human food chains e.g. killing weeds and pest using herbicides and insecticides

Microorganisms in Nutrient Cycles

- Microorganism play a vital role in nutrient cycles

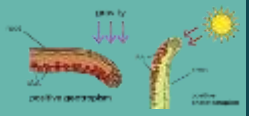
Microorganism	Role
Mycorrhizae	Certain types of fungi associate with roots of plants to increase the surface area for absorption of water and mineral ions, including phosphate ions.
Free-Living NitrogenFixing Bacteria	In the soil, they reduce nitrogen gas to ammonia.
Mutualistic NitrogenFixing Bacteria	Use nitrogen gas to produce amino acids
Saprobiontic organisms	Break down dead organism to release phosphate, ammonia or ammonium compounds
Nitrifying bacteria	Free living in soil, oxidise ammonium ions into nitrites and nitrites into nitrates
Anaerobic denitrifying bacteria	Use nitrates in respiration to produce nitrogen gas
Eutrophication	Nitrate levels increase in rivers and lakes due to leaching. The increased plant growth blocks light reaching the water underneath the surface, killing plants at a lower depth. The population of saprobiontic bacteria increase, respiring and reducing oxygen levels, killing other aerobic organisms like fish.

- Eutrophication:

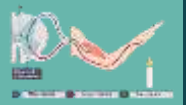


Responses

- All multicellular organisms need to respond to changes in their environment (stimuli) in order to survive
- Tropisms are a directional growth response in plants, in which the direction of the response is determined by the direction of the external stimulus
- Plants respond to directional stimuli using specific growth factors, which move to regions where they are needed from growing regions
- Indoleacetic acid (IAA) causes elongation of shoot cells, while it also inhibits root cell elongation in order to cause positive geotropism & phototropism.

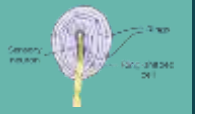


- Taxis is the movement of an animal towards or away from a stimulus
- In kinesis animals change the rate of movement (turning or speed) in order to move towards favourable conditions
- Taxis & kinesis are simple responses that can maintain a mobile organism in a favourable environment.
- Reflexes are rapid responses that don't require conscious thought.
- Reflexes can quickly protect the body from harm, as it does not involve many synapses, they use simple mechanisms and are localized to the part of the body where they occur



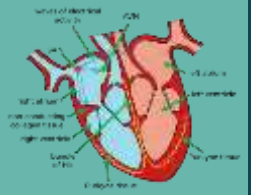
Receptors

- Sensory receptors are specialised cells in the nervous system that detect physical stimuli and convert them into electrical signals (the generator potential)
- Sensory receptors tend to be specific to one type of stimulus because they have specialised structures that are specific to one type of physical property
- Pacinian corpuscles detect changes in pressure in the skin.
- Increases in pressure cause a deformation of the concentric rings of the Pacinian corpuscle, opening stretch-mediated sodium channels in the membrane. Sodium ions enter the sensory neuron, causing a generator potential which can trigger an action potential



Control of the Heart Rate

- Cardiac muscle is myogenic, meaning it can contract and relax without receiving signals from the nervous system
- The sinoatrial node (SAN) sends out regular waves of electrical activity to the left & right atrial wall causing contraction. The electrical waves are then passed onto the atrioventricular node (AVN), then to the bundle of His, with a slight delay. The bundle of His splits into the Purkynge tissue, causing contraction of the left & right ventricles from the bottom up.

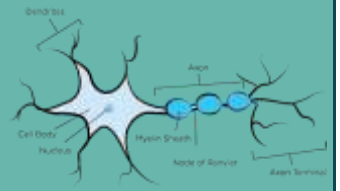


- The rate at which the SAN fires is controlled unconsciously by the medulla oblongata in the autonomic nervous system

Stimulus	Receptor	Effect
High blood pressure	Baroreceptors in the aorta & carotid arteries	Medulla sends impulses along parasympathetic neurones, using acetylcholine to reduce the heart rate
Low blood pressure		Medulla sends impulses along sympathetic neurones, using noradrenaline to increase the heart rate
High blood O_2, pH or low Co_2	Chemoreceptors in the aorta, carotid arteries & medulla	Medulla sends impulses along parasympathetic neurones, using acetylcholine to reduce the heart rate
Low blood O_2, pH or high Co_2		Medulla sends impulses along sympathetic neurones, using noradrenaline to increase the heart rate

Neurones & The Resting Potential

- A myelinated motor neurone:



- The resting potential is the difference in electrical charge across the membrane while the neurone is at rest
- The sodium-potassium pump uses ATP to pump 3 sodium (Na^+) ions out of the cell and 2 potassium (K^+) ions into the cell.

The membrane is permeable to K^+ but impermeable to Na^+ ions. These factors allow an electrochemical gradient to be set up, with the cell negatively charged at -70mV.

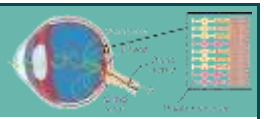
Transmission of Action Potentials

- Action potential are transmitted in non-myelinated axons because when a depolarisation happens, it causes voltagegated sodium channels to open further down the axon. By the time the depolarisation has spread, part of the axon is repolarising
- In myelinated axons, action potentials only occur at the nodes of Ranvier, with charge diffusing along the cell where myelin is present (saltatory conduction).
- Factors affecting transmission speed:

	Faster	Slower
Myelination	Myelinated	Unmyelinated
Axon Diameter	Wider	Narrower
Temperature	Warmer (Until Denaturing)	Colder

SNAPREVISE

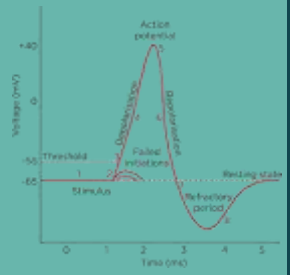
Photoreceptors



- The retina contains photoreceptors which detect light - rods and cones.

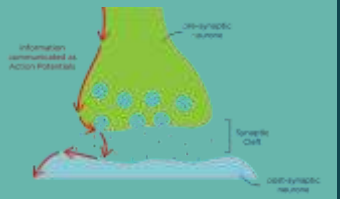
Rod cells	Cone cells
Detect light across the middle of the visible light spectrum	Three types of cone cells, which respond to red, green, and blue light
More sensitive to low light intensities than cones	Comparing the responses from each type of cone receptor allows for colour vision
Use the pigment rhodopsin to detect light	Use the pigment iodopsin to detect light
More abundant than cone cells	Fewer numbers than rod cells
Located more towards the periphery of the retina. Not present at the fovea	Concentrated at the fovea. Fewer at the periphery of the retina
Multiple rod cells connect to a single bipolar cell	Cone cells connect to their own bipolar cell
Provide poor visual acuity	Provide good visual acuity

Action Potentials



- When the neurone receives an impulse from sensory receptors, sodium channels on the dendrites open, leading to the movement of Na^+ ions into the cell causing depolarisation. If this depolarisation reaches the threshold potential it activates voltage-gated sodium channels causing an action potential. After Voltage-gated sodium ion channels close, and voltage-gated potassium channels open, causing Repolarisation as K^+ ions leave the cell. Outward diffusion of K^+ ions causes hyperpolarisation and the voltagegated potassium channels close. Finally, the Sodium-potassium pump returns the cell to the resting membrane potential.
- Action potentials are an all or nothing response because once the threshold is reached each action potential always depolarises the axon to the same voltage by voltage-gated sodium channels.
- The refractory period is the period in an action potential where the axon can't be depolarised to initiate a new action potential. It limits the frequency of action potentials and ensures action potential are discrete & only travel in one direction.

Cholinergic Synapse



- Structure of a synapse:
- At a cholinergic synapse (acetylcholine is the neurotransmitter), an action potential arrives at the pre-synaptic knob, depolarising the membrane and causes voltage-gated calcium ion channels to open. The influx of Ca^{2+} ions causes the synaptic vesicles to fuse with the membrane, releasing the neurotransmitter into the synaptic cleft. The neurotransmitter diffuses and binds receptors on the post synaptic membrane, causing an action potential.
- Acetylcholinesterase breaks down acetyl choline in the cleft.
- The synapses can be excitatory if the neurotransmitter opens Na^+ channels or inhibitory if the neurotransmitter opens chloride or potassium channels causing hyperpolarisation.
- Spatial summation is when action potentials from multiple presynaptic neurones are added together in a post-synaptic neurone
- Temporal summation is when multiple action potentials from a single presynaptic neurone are added together in a postsynaptic neurone over time.

Neuromuscular Junction

- Structure of the neuromuscular junction:
- When an action potential reaches the junction, voltage-gated calcium channels open, causing calcium ions to diffuse into the neurone. Synaptic vessels to fuse with the presynaptic membrane and release acetylcholine into the synapse. It diffuses across the synapse and binds with receptors on the muscle cell surface membrane, opening sodium channels. The muscle fibre depolarisation causes an action potential and muscle contraction.
- Acetylcholinesterase breaks down acetyl choline
- Neuromuscular junction & cholinergic synapse differences

Neuromuscular Junction	Cholinergic Synapse
Only excitatory	Can be excitatory or inhibitory

Homeostasis

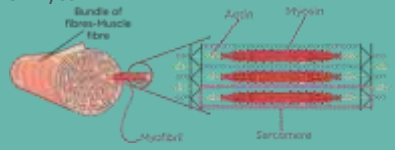
- Homeostasis is the maintenance of a constant internal environment despite internal or external changes.
- Temperature & pH are important to regulate to allow optimum enzyme activity and rate of metabolic reactions.
- Water potential is important to regulate to prevent cells bursting or shrinking.
- Glucose concentration is important to regulate to allow cells to have access to the substrate for respiration, whilst preventing cell damage by dehydration caused by high concentrations.
- Negative feedback is the body's mechanism for reversing a change so that it returns back to the optimum. The stages involve:

- Positive feedback is a deviation from the optimum which causes changes resulting in an even greater deviation from the norm. This is usually harmful due to the large, unstable change in the body.

Links neurones to muscle	Links either neurones to neurones or neurones to other effectors
The action potential ends here	Another action potential may be generated along the postsynaptic neurones
Only motor neurones are involved	Intermediate, motor and sensory neurones may be involved
Acetylcholine binds to receptors on the membrane of the muscle fibre	Acetylcholine binds to receptors on membrane of post-synaptic neurone

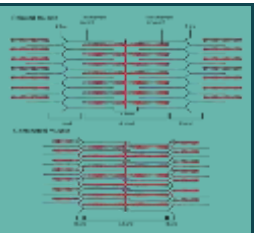
Skeletal Muscles

- Muscles act in antagonistic pairs against an incompressible skeleton to allow movement
- Skeletal muscle is made up of fibres called myofibrils, which in turn are made up of many repeating units, called sarcomeres
- Myofibrils are made up of two types of protein filaments, the thinner actin and the thicker myosin



	Slow-Twitch Muscle	Fast-Twitch Muscle
Type of Activity	Endurance	Burst of activity

Muscle Contraction



- The sliding filament theory describes how muscle contraction occurs
- An action potential travels into the muscle fibre via T tubules, causing release of calcium ions from the sarcoplasmic reticulum. The calcium ions bind to the tropomyosin molecules and cause them to move, exposing the myosin binding site on the actin filament. Myosin attaches to actin forming a actin-myosin cross-bridge. ATPases hydrolyse ATP to detach the myosin head, allowing reattachment at a further site. This cycle continues, causing sarcomeres to shorten.
- When nervous stimulation stops, Ca^{2+} ions are actively transported back into the sarcoplasmic reticulum using energy from ATP hydrolysis. This allows tropomyosin to block the actin filament from binding to myosin and muscle contraction stops.
- ATP can be generation via aerobic or anaerobic respiration
- Phosphocreatine generates ATP quickly by adding phosphate to a molecule of ADP released by the contracting muscle

Communication Systems

- The neuronal system uses neurones to carry signals very rapidly through the body to produce short-term responses
- The hormonal system uses blood to carry hormones from endocrine glands to target cell with the specific receptors. This usually produces long-term responses.
  - Peptide hormones are made of amino acids and must bind to receptors on the cell surface, activating second messengers which control transcription.
  - Steroid hormones are formed from lipids and soluble in the plasma membrane, therefore entering cells and binding to proteins to enter the nucleus and have an effect on the DNA.

Diabetes

- Diabetes is a condition where the concentration of glucose in the blood cannot be controlled effectively. It can lead to hyperglycaemia after meals and hypoglycaemia after exercising.
- Type 1 diabetes is caused by an autoimmune attack on the β-cells of the pancreas, so the body cannot produce insulin. It can be treated by insulin injections.
- Type 2 diabetes is caused because the body does not produce enough insulin & the insulin receptors become less responsive. It can be treated by lifestyle changes (losing weight & exercising), drugs to stimulate insulin production and reduce glucose absorption and insulin injections in severe cases.

Control of Blood Glucose Concentration

- Insulin is a hormone released from β-cells in the pancreas when blood glucose concentration rises in order to lower the concentration back to its optimum via negative feedback.
- When insulin binds to an insulin receptor, vesicles of glucose transporters fuse with the plasma membrane to allow more glucose to enter the cell. The cell also uses more glucose in respiration and activated enzymes covert glucose into glycogen (glycogenesis).
- Glucagon is a hormone released from α-cells in the pancreas in response to low glucose concentration in order to increase the concentration back to its optimum. It does this by:
  - Activating enzymes which break down glycogen into glucose (glycogenesis).
  - Producing glucose from other molecules
  - Activating enzymes that convert glycerol (from lipids) and amino acids into glucose (gluconeogenesis)
- Adrenaline is released by the adrenal glands in times of stress and increases blood glucose concentration in anticipation of increased activity.
- Adrenaline binds to adrenaline receptors which activates adenyl cyclase. This converts ATP into cAMP, which acts as a second messenger to activate protein kinase. Protein kinase converts glycogen into glucose.

Contraction Details	Contracts slowly and for longer Fatigues slowly	Contracts quickly and then relaxes rapidly
Mitochondria Density	High	Low
Type of Respiration	Aerobic	Anaerobic
Concentration of Myoglobin	High concentration	Low concentration
Glycogen & Phosphocreatine Stores	Small	Large
Muscle Colour	Dark	Light

The Role of the Hypothalamus in Osmoregulation

- The hypothalamus contains osmoreceptors which signal to specialised neurosecretory cells. A fall in water potential causes the release of antidiuretic hormone (ADH) from the pituitary gland.
- ADH travels in the blood to the kidneys, attaching to ADH receptors, activating the intracellular enzyme phosphorylase. This causes vesicles containing aquaporins to fuse with the plasma membrane, reducing water loss by increasing the permeability of the collecting duct and distal convoluted tubule.

The Role of the Kidneys in Osmoregulation

- Osmoregulation is maintaining a constant water potential of the blood, despite changes in the level of water and salt intake.
- The kidneys are made if nephrons which help filter the blood. The blood undergoes ultrafiltration at the glomerulus due to the smaller diameter of the efferent arteriole than the afferent arteriole, creating high hydrostatic pressure.

- The filtrate passes into the Bowman's capsule around the entire nephron, where certain water are reabsorbed into the blood whilst remaining filtrate is excreted as urine.
- Sodium is actively transported out of the proximal convoluted tubule and into the blood
- Glucose & amino acids are co-transported out of the proximal convoluted tubule via sodium ions diffusing into the epithelial cells.

and travels ions and the

Epistasis

- Epistasis is the interaction between two non-linked genes which causes one gene to mask the expression of the other in the phenotype.
- Epistatic genes can work antagonistically (against each other) or in a complementary fashion.
- When a gene suppresses another gene, the gene doing the suppressing is called the epistatic gene. The gene which is being suppressed is called the hypostatic gene.
- Antagonistic epistasis can be either recessive or dominant.
- In dominant antagonistic epistasis, the expression of the dominant allele of the epistatic gene prevents the expression of the hypostatic gene. This means that any genotypic combination with either one or two of the dominant alleles for the epistatic gene will suppress the expression of the hypostatic gene.
- Recessive epistasis occurs when the presence of two copies of the recessive allele at the first locus prevents the expression of another allele at a second locus.
- In complementary epistasis, the two genes work together, for example, they may encode two enzymes that work in succession.

Chi-squared Test

- If during an experiment, an unexpected result is obtained, we need to determine whether this unexpected result is due to chance or attributable to a specific cause (significant or not).
- The chi-squared test is a type of statistical test that allows us to calculate whether the difference between the results we observe and the results we expected is significant.
- The null hypothesis assumes that any difference that occurs between the expected and observed results is due to chance.

Chi-Squared Test

O is the observed numbers (no units)

E is the expected numbers (no units)

- The value is then compared to a critical value, found from a chi-squared table by looking at the p-value and degrees of freedom
  - The degrees of freedom is the number of categories (or classes) minus one
  - The *p*-value is normally taken as 0.05, meaning that there is a 5% probability that the result is due to chance only
- If < critical value, then the results are not significant (are due to chance). The null hypothesis is accepted.
- If > critical value, then the results are significant (are attributable to a specific cause). The null hypothesis is rejected.

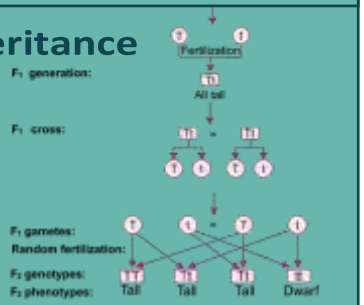
SNAPREVISE

Genes & Alleles

- The genotype is an organism's genetic composition.
- The phenotype is an organism's characteristics, often visible, which occur as a result of both its genotype and the impact of its environment.
- Genes are a sequence of DNA that code for a polypeptide.
- Genes can exist in 2 or more different forms called alleles.
- In diploid cells, chromosomes occur in pairs called homologous chromosomes. This means the alleles at a specific locus can be homozygous if they are both the same type of allele or heterozygous, if both the alleles are different.
- An allele is dominant if it is expressed in the phenotype of an heterozygous individual.
- An allele is recessive if it is not expressed in the phenotype of an heterozygous individual.
- An allele is codominant if it is expressed, along with the other allele, in the phenotype of a heterozygous individual.

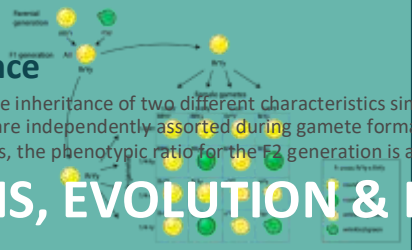
Monohybrid Inheritance

- Monohybrid inheritance is the inheritance of a single gene.
- A test cross be used to work out the unknown genotypes of individual organisms.
- In the test cross the unknown genotype is crossed with a homozygous recessive individual. If all the offspring have the dominant phenotype, the unknown genotype was homozygous dominant for the trait. If half the offspring have the recessive phenotype, the unknown genotype was heterozygous.



Dihybrid Inheritance

- Dihybrid inheritance involves the inheritance of two different characteristics simultaneously.
- During a dihybrid cross, alleles are independently assorted during gamete formation. A punnet square can show all possible genotype and phenotypes of offspring:
- In a dihybrid F1 generation cross, the phenotypic ratio for the F2 generation is always 9:3:3:1.

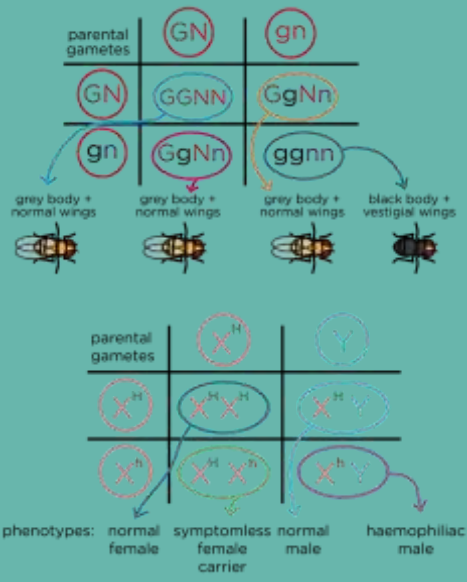


POPULATIONS, EVOLUTION &

SUMMARY SHEET

Linkage

- Autosomal linkage occurs if two or more genes are located on the same autosome (non-sex chromosome). The two genes are less likely to be separated during crossing over, resulting in the alleles of the linked genes being inherited together.
- For example, if GN & gn are linked in heterozygous grey bodies and normal winged individuals (GgNn), you get a 3:1 phenotypic ratio
- Sex linkage occurs when there is a gene on the X chromosome, not present on the Y chromosome.
- This means that males are more likely to exhibit recessive disorders like haemophilia



Complementary Epistasis Example

- An example of complementary epistasis is in the inheritance of coat colour in mice.
  A/a is the epistatic gene
  AA & Aa produces coloured fur
  aa produces no pigment-white fur
  B/b is the hypostatic gene
  BB & Bb encodes for black coloured fur
  bb produces encodes for agouti coloured fur

AaBb X AaBb

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

This produces a 9:4:3 phenotypic ratio

SNAPREVISE

Populations

- A species is a is a group of individuals that have common ancestry and are capable of breeding with each other and producing fertile offspring.
- Species exist as one or more populations
- A population is a group of organisms of the same species occupying a particular space at a particular time that can potentially interbreed.
- A gene pool is all of the alleles of all the genes of all the individuals of a population
- Allele frequency is the proportion of the individuals that have one copy of an allele
- Allele frequencies change in response to selection pressures by natural selection between and within populations.

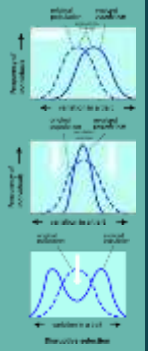
Population Genetics

- Populations can be imagined as gene pools consisting of all the alleles of all the genes of all the individuals in the population
- Populations change and evolve as allele frequencies change across generations
- The frequency of alleles of a particular gene in a population can be determined using the equation encompassed by the HardyWeinberg Principle
- Hardy-Weinberg equations: $p + q = 1$

  $p2 + 2pq + q2 = 1$
- Where:
  p is the frequency of dominant allele q is the frequency of recessive allele
  p^2 is the proportion of individuals that are homozygous dominant (AA) q^2 is the proportion of individuals that are homozygous recessive (aa)
  2pq is the proportion of individuals that are heterozygous (Aa)
- Using the equations, the allele frequencies of a specific gene, genotypes & phenotypes in a population can be estimate.
- The Hardy-Weinberg Principle assumes that the proportion of dominant and recessive alleles of any gene in a population remains the same from one generation to the next. The conditions for this are that:
  - The population is large
  - There are no mutations
  - There is no selection
  - Mating is random within the population
  - The population is isolated

The Effect of Selection on Allele Frequencies

- Predation, disease and competition means that not all individuals within a population survive to get a chance to reproduce. This differential survival and reproduction is the process by which natural selection acts.
- The organisms with phenotypes that provides a selective advantage are more likely to reproduce and thus pass on their favourable alleles to the next generation. This means that the proportional of individuals with the favourable allele will increase in the next generation (increase the allele frequency) within the population. The population evolves.



- Evolution is the change in allele frequencies in a population over time.
- Directional selection results in the increase of a favoured allele over time
- Stabilising selection maintains genetic polymorphisms in the population
- Disruptive selection also maintains genetic polymorphisms in the population

Speciation

- Speciation is the evolution of new species from existing ones.
- Reproductive isolation followed by accumulation of genetic changes through natural selection can result in the formation of a new species. This is because the populations become genetically distinct with different allele combinations, making them unable to breed to produce fertile offspring.

- Allopatric speciation is the formation of two species from an original one due to geographical isolation.
- Sympatric speciation is the formation of two species from one original species due to reproductive isolation whilst occupying the same geographical location. This can be by:
  - Temporal variation - breeding seasons at different times.
  - Behavioural variation - mutations affecting courtship.
  - Mechanical variation - anatomical differences preventing mating.
  - Gametic variation - results in genetic or biochemical incompatibility.
  - Hybrid sterility - cannot produce viable gametes.

Ecosystems & Population Size

- A community is all of the populations of different species living and interacting in a place at the same time.
- An ecosystem is the dynamic interaction between all the living (biotic) and non-living (abiotic) factors in a given area.
- Within an ecosystem, every organism occupies a specific ecological niche
- A niche includes all the abiotic and biotic conditions of the environment which organisms are adapted to.
- The carrying capacity is the maximum population size that can be maintained over a period in a particular habitat.
- The limiting factors of the carrying capacity include abiotic factors:
  - Temperature & pH - each species has its optimum levels, and deviations from this optimum reduces population growth
  - Light - low light levels reduce the carrying capacity of producers, reducing the population size of consumers
  - Water - low water availability reduces the population size
- The limiting factors of the carrying capacity include biotic factors:
  - Interspecific competition (between different species)
  - Intraspecific competition (within the same species) ◦ Predation
- The size of a population can be estimated by:
  - Randomly placing quadrats, or quadrats along a belt transect, for slow-moving or non-motile organisms. Can count the number of individuals of each species in the quadrat or percentage cover.
  - The mark-release-recapture method for motile organisms.

$$Population\ size = \frac{sample\ 1\ size \times sample\ 2\ size}{\#\ of\ marked\ in\ sample\ 2}$$

It assumes there is no deaths, births, migration, marking has no effect and enough time for the animals to mix.

Succession

- Succession is the variety of processes that occur over time in a species that occupy a certain area.
- Primary succession is the progressive colonisation of bare rock or other barren terrain by living organisms.
  - The area is first colonised by the pioneer species, changing the abiotic factors to be less hostile for other species to survive.
  - Different species may be present at each stage, who change the environment so that it becomes more suitable for other species with different adaptations but less suitable for the previous species - changing biodiversity.
  - The climax community is when a stable state is reached, where there is high biodiversity and a number of new species.
- Secondary succession is the recolonization of an area after an earlier community has been removed or destroyed.

SNAPREVISE

Variation

- Within any population of a species there will be phenotypic variation
- Characteristics that show continuous variation are normally polygenic (determined by many gene loci that have additive effects on each other).
- Characteristics that show discontinuous variation are usually monogenic (determined by a single gene loci).
- Variation is due to genetic and environmental factors.
- The main source of genetic variation is mutations, which can produce different alleles of genes.
- Further sources of genetic variation include meiosis (independent assortment and crossing over) and the random fertilisation of gametes during sexual reproduction to create new allele combinations.
- The environment can influence the way an organism's genes are expressed. This can be because of biological factors such as predators or non-biological factors such as sunlight.

Genetic Drift

- Genetic drift describes change in allele frequencies in the gene pool of a population (evolution) due purely to chance events and not selection pressures.
- Due to the random nature of gamete production and fertilisation, certain alleles may increase in the population due to chance.
- The effect of genetic drift is more prominent within small populations because chance has a greater influence, whereas in larger populations the random fluctuations even out across the whole population.
- A genetic bottleneck is when an event causes a big reduction in a population's size and gene pool. Certain alleles may be due to the event and the population will also be subject to genetic drift.
- When a new population is established by a small number of individuals, the founding population will have low genetic diversity and be heavily influenced by genetic drift. This is the founder effect.

Conservation

- Conversations is the maintenance of biodiversity, including diversity between species, genetic diversity within species and maintenance of a variety of habitats and ecosystems.
- Conservation involves active human involvement and is often orientated around managing a community by halting succession, to preserve species that would be extinct by the climax community being established.
- The rate of growth of the human population creates an increasing demand for raw material and food. A balance between conversation and human needs is necessary in order to maintain the sustainability of natural resources.

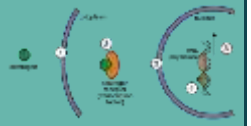
Mutations

- Gene mutations are changes to the base sequence or quantity of DNA within a gene or section of DNA.
- Gene mutations occur spontaneously during the process of DNA replication.
- The mutation rate is increased by mutagenic agents, which are chemical, physical or biological agent that causes mutations e.g. UV light

Type of Mutation	Description
Addition	Addition of one or more nucleotides
Deletion	Removal of one or more nucleotides
Substitution	A nucleotide is replaced by a different nucleotide
Inversion	A sequence of bases is separated and then reattached in the inverse order
Duplication	One or multiple bases are repeated
Translocation	A piece of DNA breaks off and doesn't reattach to itself or its homologous pair.

- Some mutations may only affect a single codon, changing a single amino acid in a protein, therefore the protein may remain functional. Other may have no effect on protein structure due to the genetic code being degenerate.
- Mutations such as insertions and deletions can cause frame shifts, changing all the codons and amino acids downstream from the mutation. This results in a unfunctional protein.

Transcription Factors



- In eukaryotes, transcription of target genes can be regulated by DNA-binding proteins (transcription factors). They can be help RNA polymerase bind (activators) or prevent it binding (repressors).
- The steroid hormone oestrogen, released from the ovaries in women, can initiate transcription in target cells.

Epigenetics

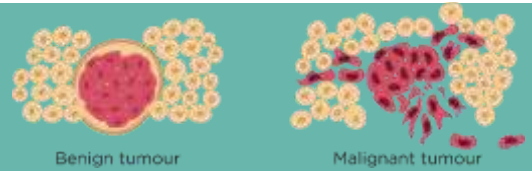
- Epigenetics - changes in DNA that alter the expression of genes without changing the base sequence of DNA itself. It involves the addition of chemical tags onto DNA or histones.
- The epigenetic changes can regulate transcription by changing how tightly the chromatin is packed (chromatin remodelling), affecting RNA polymerase accessibility.

- DNA methylation prevents transcription by preventing transcription factors from binding & chromatin condensation.
- Acetylation of histones promotes transcription by decreasing the attraction between DNA and histones, making chromatin more loosely packed.
- The epigenetic changes in gene function can be heritable.
- Epigenetic changes occur during development but can also be caused by environmental factors e.g. smoking.

Tumours

- Abnormal and fast cell division of mutant cells can form a tumour.

Benign Tumours	Malignant Tumours
Slow growth rate	Faster growth rate
Cells remain welldifferentiated	Cells tend to de-differentiate and become unspecialised
Tumours are surrounded by a capsule made of dense tissue (compact structure)	Tumours are not surrounded by a capsule
Cells produce adhesion molecules	Cells stop producing adhesion molecule. Can spread through the body (metastasis)
Can usually be removed by surgery.	Chemotherapy and radiotherapy are used, which specifically target and kill rapidly dividing cells.



Stem Cells

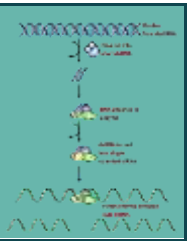
- Stem cells are undifferentiated cells that are able to express all of their genes and divide by mitosis.
- During development, the stem cells undergo cell differentiation. This is the process by which cells become specialised for different functions.
- Fully developed cells are unable to divide by mitosis.

Stem Cell	Ability
Totipotent	Can divide and differentiate into any type of cell.
Pluripotent	Can self-renew and differentiate into any type of cell except the cells that make up the placenta.
Multipotent	Can only differentiate and divide into a limited number of cell types
Unipotent	Can only differentiate into a single type of cell e.g. cardiomyoblasts can only differentiate into cardiomyocytes.

- Totipotent stem cells are only present in mammals in the first few cell divisions of an embryo. During development, totipotent cells become specialised by expressing different genes and producing different proteins.
- Induced pluripotent stem cells are unipotent stem cells that have been reprogrammed to become pluripotent by using protein transcription factors to express genes associated with pluripotency.
- Pluripotent stem cells can be used to replace cells and treat human disorders like leukaemia and diabetes.

Regulating Translation

- In eukaryotes and prokaryotes, the translation of mRNA can be inhibited by RNA interference (RNAi).
- RNAi involves the degradation of the mRNA, reducing the gene's level of expression. Small interfering RNA (siRNA) can carry out this process.

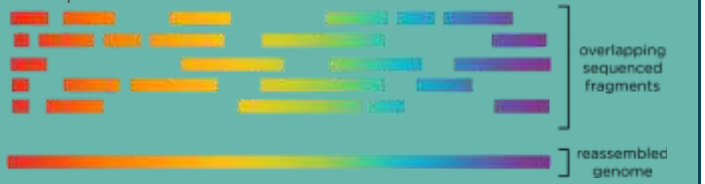


Oncogenes & Tumour Suppressor Genes

- Oncogenes are genes that stimulate cell division e.g. they may encode growth factors or cell cycle regulators.
- Many cancers are found to have cells with abnormal DNA methylation (epigenetic changes). Detecting these changes can help diagnose, while reversing these changes may help cure these diseases.
- Oncogenes can be hypomethylated in the promoter regions to upregulate transcription and expression to cause excessive proliferation in a tumour.
- Tumour suppressor genes are genes that prevent tumour formation by repairing DNA damage, regulating cell division and promoting apoptosis.
- Tumour suppressor genes can be hypermethylated in the promoter region to prevent transcription, allowing increased cell divisions with a higher mutation rate. Resulting in cancerous tumours.
- Oestrogen binds to a transcription factor, which activates genes to promote cell division. Increased oestrogen concentrations in the adipose tissue in the breast of post-menopausal women has been linked to breast cancer development.

Genome Projects

- DNA sequencing is the process used to determine the precise sequence of nucleotides in a length of DNA.
- The technique whole-genome shotgun sequencing is used. The genome is cut into smaller fragments and individually sequenced. The entire genome is then reassembled by computer algorithms, which align sections of DNA that overlap.



- Next-generation sequencing methods have recently been developed which are faster, more automated and cheaper.
- Whole-genome sequencing allows the genomes of many individuals within a species, to be compared.
- This can have important medical implications by looking for associations between substitution mutations (single nucleotide polymorphisms, SNPs) and susceptibility to disease.
- In simpler organisms, such as pathogens, genome sequencing allows the proteome to be determined. This can help determine potential cell surface proteins that act as antigens, which can be used in vaccine development.
- In more complex organisms, determining the proteome is more difficult due to the presence of introns, regulatory genes affecting the expression of other genes & the effect of epigenetic changes.

Genetic Engineering

- Genetically modified organisms are organisms that have had their DNA altered through recombinant DNA technology.
- Recombinant DNA technology involves the transfer of fragments of DNA from one organism, or species, to another.
- Transgenic organisms can successfully express a gene from any organism, as the genetic code and mechanism of protein production (transcription and translation) are universal.
- DNA fragments are created by:
  - Using restriction endonucleases to cut at recognition sites near the desired gene
  - Converting the mRNA of the desired gene to cDNA, using reverse transcriptase. Double stranded DNA is then synthesised using DNA polymerase
  - Synthesising the gene using a gene machine. The gene sequence is determined by the primary protein structure.
- The isolated gene is then modified by the addition of a promoter and a terminator region.
- A vector is used to transfer the isolated gene into a host cell. This is mainly a plasmid.
- Restriction endonucleases are used to cut plasmids open, creating sticky ends. The same endonuclease isolates the gene, so the sticky ends of the desired gene and the plasmid are complementary. DNA ligase joins them together.
- To reintroduce the desired DNA into bacterial cells, the recombinant plasmid must pass through the cell surface membrane of a bacterial cell (transformation).
- Transformation involved mixing the bacteria and plasmids in a medium containing Ca^{2+} ions, which increased membrane permeability. Changes in temperature also make the bacterial cell surface more permeable.
- The transformed host cells can be cultured as an in vivo method to amplify DNA fragments.

Marker Genes

- Transformed bacteria can be detected using marker genes.
- The plasmid contains 2 marker genes
  - The first marker gene is used to identify which bacteria have successfully taken up a plasmid. It is a antibiotic resistance gene, so transformed bacteria are identified by growing on a medium containing the antibiotic
  - The second marker distinguishes between bacteria that have taken up an empty or recombinant plasmid. When a recombinant plasmid is formed, the desired gene is inserted in the middle of the second marker gene making it nonfunctional. Therefore, bacterial cells that express the second marker gene do not contain the recombinant plasmid.
- The second marker gene has easily identifiable phenotypes such as:
  - Producing a fluorescent protein
  - Providing resistance to a different antibiotic
  - Producing an enzyme whose action can be identified.

Polymerase Chain Reaction (PCR)

- PCR is a method of amplifying DNA by artificial replication in vitro.
- It requires: DNA sample of around 10,000 base pairs, nucleotides, Taq polymerase (stable at high temperatures), primers complementary to 3' of DNA sample and a thermocycler to carry out the automated process.

Gene Therapy

- Gene therapy is the mechanism by which genetic diseases are treated or cured by masking the effect of a faulty allele through the insertion of a functional allele.
- Firstly, a healthy allele from healthy cell tissue is isolated. The allele is inserted into the cells using vectors.
- If the mutated allele is recessive, a dominant allele is inserted. If the mutated allele is dominant, DNA is inserted into the middle of the allele to silence it.
- Somatic therapy involves altering the alleles in body cells. The altered allele is not passed onto the offspring
- Germ-line therapy altering the alleles in the sex cells. The altered alleles are passed onto offspring
- Germ-line therapy has ethical concerns such as the potential of designer babies or the potential impact gene insertion could have on other genes.

Diagnosing Heritable Conditions

- Genetic screening is the study of an individual's DNA to identify whether an individual possesses alleles associated with a genetic disease.
- Genetic screening can be carried out using DNA probes which are short sections of DNA that are complementary to a known DNA sequence (e.g. a mutant allele). The probes are labelled using fluorescence or radioactivity.
- The labelled DNA probe, which is complementary to a mutant allele, is mixed with denatured DNA samples from a patient. If the patient has the mutant allele, the probe will bind to the complementary base sequence in one strand (hybridization). The hybridized DNA can be detected using radiation or fluorescence.
- DNA probes can be used to screen patient for different genetic diseases, to see if they are carriers for a recessive mutation or to see if they are at risk of developing a disease like cancer, by having mutated oncogenes or tumour suppressor genes.
- Genetic screening also allows medicine or treatments to be precisely tailored to an individual's genotype (personalised medicine).
- After receiving the results of genetic screening, individuals may require genetic counselling. This is a service that provides support, information and advice about genetic conditions.

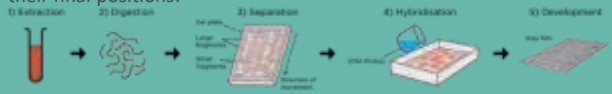
The Use of Genetically Modified Organisms (GMOs)

GMO	Benefits	Issues
Plants	• Herbicide resistance • Pest resistance • Disease resistance • Drought resistance • Extended shelf-life • Increased nutrition	• Development of superweeds • Pests or pathogens evolving resistance • Potential transfer of antibiotic resistance to pathogens in the intestine of the consumer • Farmers must repeatedly buy seeds
Animals	• Disease resistance • Increased growth rates e.g. continuously producing growth hormones • Used to produce medicinal drugs and proteins	• Harmful side effect to animals • Ethical issue of insertion of human genes • Most GM animals die during development
Bacteria	• Used to produce medicine e.g. human insulin which is cheaper and has a lower risk of rejection and infection than pig insulin	• Potential antibiotic resistance genes being transferred to pathogens • May result in the production of more lethal pathogens

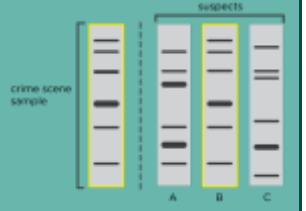
- The risk of GM bacteria can be reduced by modifying the bacteria so that they are unable to produce an essential nutrient or amino acid and cannot survive outside the lab.

Genetic Fingerprinting

- Genetic fingerprinting is a method used to produce a specific pattern of DNA bands from an individual's genome.
- The non-coding regions of DNA contain short, repeating sequences called variable number tandem repeats (VNTRs).
- VNTRs are found at many locations in the genome. In every individual, they vary in length and the in the number of repeats at different loci. Therefore, the probability of two individuals having the same VNTRs is very low.
- The steps in DNA fingerprinting include:
  - Extraction of DNA & amplification using PCR
  - DNA digestion using specific restriction endonucleases, leaving the VNTRs intact
  - Separation of DNA fragment by gel electrophoresis. Smaller fragments travel faster and therefore move further down the gel
  - Hybridisation of the VNTRs at specific (complementary) base sequences with Radioactive or fluorescent DNA probes
  - Development. The banding pattern can then be visualised as radiation, emitted by fragments, exposes X-ray film (placed over the gel) and reveals their final positions.



- The DNA profiles can be compared to determine genetic relationships by looking for similarities in the banding pattern.
- DNA profiles can also be used in:
  - Forensic science investigations - comparing the DNA profiles of suspects and DNA at the crime scene.
  - Medical diagnosis - DNA profiles can identify individuals at risk of developing specific diseases, as some VNTRs are correlated with an increased risk of disease e.g. Huntington's disease.



  - Animal and plant breeding - DNA profiles are used to prevent inbreeding by not breeding individuals with similar profiles.
  - Paternity determination - half the DNA profile of the child should match the father.