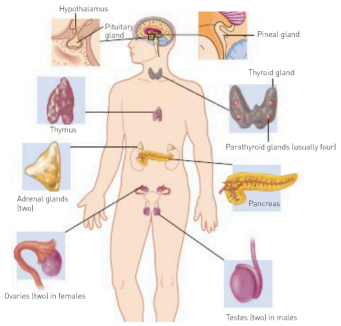
HUMAN BIOLOGY TOPIC 1

THE ENDOCRINE SYSTEM



The endocrine system is composed of a group of ductless glands that secrete hormones into the bloodstream. These aid the body in homeostatic control as well as the nervous system.

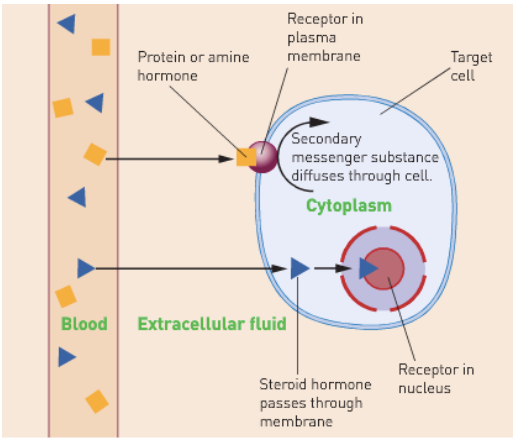
HORMONES

Hormones be proteins, steroids or amines. They are transported in the blood, affecting target cells/organs. Cells may communicate with each other in the same tissue by secreting local hormones or **paracrines.** They work slowly and are secreted by **endocrine glands.**

Hormones have **specific receptors** much like enzymes. If occupied, receptors do not have a greater affect with the addition of more hormones.

TYPES OF HORMONES

Lipid soluble (steroid proteins)
- Able to diffuse through the cell membrane
- Binds to receptor inside the cell
- Activates hormone/receptor complex to alter gene expression and cell metabolism



Water soluble (protein/amine hormones)
- Not able to diffuse through the cell membrane
- Attaches to a receptor on the cell membrane
- Secondary messenger is sent to a receptor inside the cell
- Alters gene expression and cell metabolism

HORMONE FUNCTION

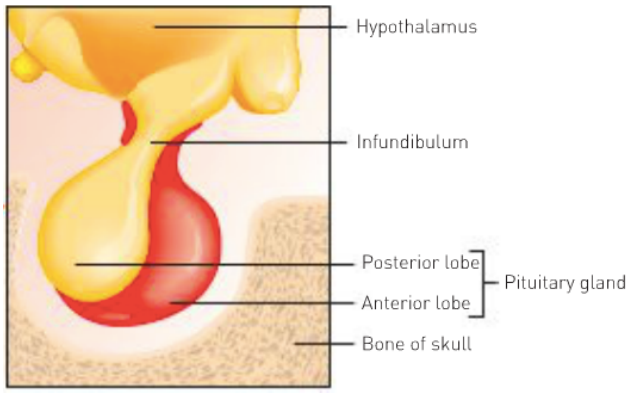
Hormones change the function of cells by changing the type, activities and quantities of proteins.
- They activate certain genes to produce specific enzymes or structural proteins
- They may change structure or shape of enzyme (turn on/off)
- They also change rate of production of an enzyme or structural protein via changing transcription/translation rate

Hormones are excreted in bile and urine and are broken down in the liver or kidney.

HYPOTHALAMUS AND PITUITARY GLAND

The **hypothalamus** is located in the base of the brain and regulates many basic functions such as body temperature, water balance and heart rate.

- Hypothalamus produces releasing factors that **stimulate** the production of a hormone in anterior pituitary gland
- May also **inhibit** the production of hormones
- Posterior lobe does not produce their own hormones, produced by hypothalamus
- Connected to pituitary gland via infundibulum



ANTERIOR LOBE OF PITUITARY GLAND

Secretions of the anterior pituitary gland are controlled by releasing inhibiting factors secreted by the hypothalamus into the extracellular fluid. True endocrine gland as it produces its own hormones.

HORMONE	TARGET ORGAN	EFFECTS
FOLLICLE STIMUATING HORMONE (FSH)	- Ovaries - Testes	- Growth of Follicles - Sperm production
LUITENISING HORMONE (LH)	- Ovaries - Testes	- Ovulation and corpus leutem formation - Secretion of testosterone
GROWTH HORMONE (GH)	All cells	Growth and synthesis
THYROID STIMULATING HORMONE (TSH)	Thyroid gland	Secretes hormones such as thyroxine from thyroid
ADRENOCORTICOTROPHIC HORMONE (ACTH)	Adrenal Cortex	Secretes hormones such as cortisol from adrenal cortex
PROLACTIN (PRL)	Mammary Glands	Milk production

POSTERIOR LOBE OF PITUITARY GLAND

Is not a "true endocrine gland" as it does not produce either hormone that it secretes. They're produced in special nerve cells in the hypothalamus called **neureosecretory cells.**

HORMONE	TARGET ORGAN	EFFECTS
ANTIDIURETIC HORMONE (ADH)	Kidneys	Increase the reabsorption of water
OXYTOCIN (OT)	- Uterus - Mammary Glands	- Uterine contractions - Release of milk

ENDOCRINE GLANDS

PINEAL GLAND
- found deep inside brain
- Secretes hormone **melatonin** which is involved with regulating sleep

THYROID GLAND
- located in neck and consists of two lobes
- Secretes **thyroxine** which is made of iodine and amino acids
- Controls body's metabolism by increasing metabolic rate
- Releases energy and maintains body temperature
- Stimulated by TSH

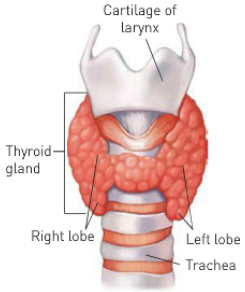
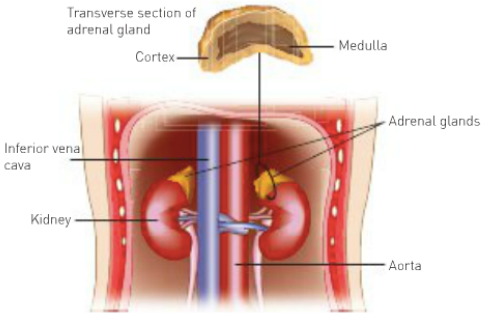


Figure 2.5 The location of the thyroid gland

PARATHYROID GLAND
- Found in lobes of thyroid gland
- Secrets **parathyroid hormone** which controls calcium and phosphate levels in blood

THYMUS
- Located just above the heart
- Secretes group of hormones called **thymosins**
- Influence the maturation of T-lymphocytes

ADRENAL MEDULLA
- Produces **adrenaline** which helps prepare the body to react in threatening situations
- Also produces **noradrenaline** which which increases heart rate ad force of heartbeat
- Increases metabolic rate of cells which increases production of heat



ADRENAL CORTEX
- Produces more than 20 different hormones, collectively called **corticosteroids**
- **Aldosterone** reduces sodium and increases potassium in urine
- **Cortisol** promotes normal metabolism, helping the body withstand stress and repair damaged tissues

THE PANCREAS
- The endocrine part secretes digestive enzymes into small intestine
- Pancreatic Islets secrete **insulin** which stimulates the uptake of glucose from the blood into cells, reducing blood glucose levels
- **Glucagon** increases the blood sugar levels by breaking down glycogen to glucose

THE GONADS
- **Androgens** in the testes develop/maintain male sex characteristics
- **Oestrogen** and progesterone in the ovaries maintain female sex characteristics and regulate menstruation and pregnancy

THE NERVOUS SYSTEM

The nervous system is the communication system and control centre of the body. With the endocrine system, it maintains a constant internal environment within the body. It is composed of two sections; the **Central Nervous System (CNS)** which is made up of the brain and spinal cord and the **Peripheral Nervous System (PNS)**

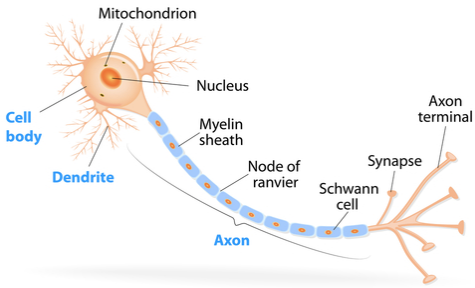
NEURON STRUCTURE

Neurons are the basic structural and functional units of the whole nervous system. They vary is shape and size.

- All neurons consist of a cell body and an extension from the cell (called the **dendrites and axons**)
- Have many branches that send and receive messages
- Most axons are covered in a layer of fatty material called the **myelin sheath**
- Nerve fibres are any long extensions from the neurons
- Those that have a myelin sheath are **myelinated fibres** and those that don't are **unmyelinated**
- **Grey matter** consists if nerve cell bodies and unmyelinated fibres
- **White matter** is composed of myelinated fibres
- **Schwann cells** wrap around the axons to form the myelinated sheath
- **Nodes of Ranvier** are the intervals along the axon
- **Neurilemma** help repair injured fibres and are the thin layer outside the Schwann cell

The role of the Myelin Sheath:

1. Acts as an **insulator**
2. **Protects** the axon from **damage**
3. **Speeds up** movement of nerve impulses along the axon

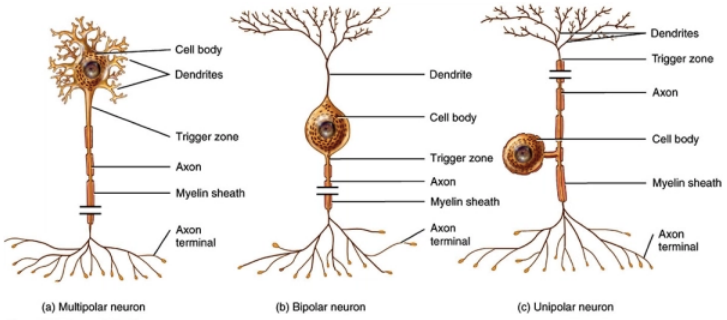


FUNCTIONAL TYPES OF NEURONS

Sensory/receptor neurons - carry messages from receptors in the sense organs to the CNS
Motor/effector neurons - carry messages to the muscles and glands from the CNS
Interneurons/connector neurons/association neurons - located in the CNS and link between sensory and motor neurons

STRUCTURAL TYPES OF NEURONS

Multipolar neurons - have one axon and multiple dendrites. Its the most common type and includes most of the interneurons in CNS and motor neurones
Bipolar neurons - have one axon and one dendrite and occur in places such as the ear, eye and nose
Unipolar neurons - have just one extension, an axon, and include most sensory neurons



SYNAPSES

Synapses are nerve impulses that have been passed from one neuron to another. Neurons don't join at the synapse, but instead messages are carried across the small gap via **neurotransmitters.** This gap is referred to as the **neuromuscular junction.**

This process occurs only in **one direction**, from axon to dendrite. It occurs in a series of steps:

1. CALCIUM CHANNELS OPEN
- incoming action potential causes depolarisation in the **presynaptic knob**
- **Voltage-gated** sodium channels open
- Calcium flood into the presynaptic knob

2. NEUROTRANSMITTER RELEASE
- Influx of calcium ions cause the synaptic vesicles to move and fuse with the presynaptic membrane
- Neurotransmitters released into the synoptic cleft by **exocytosis**

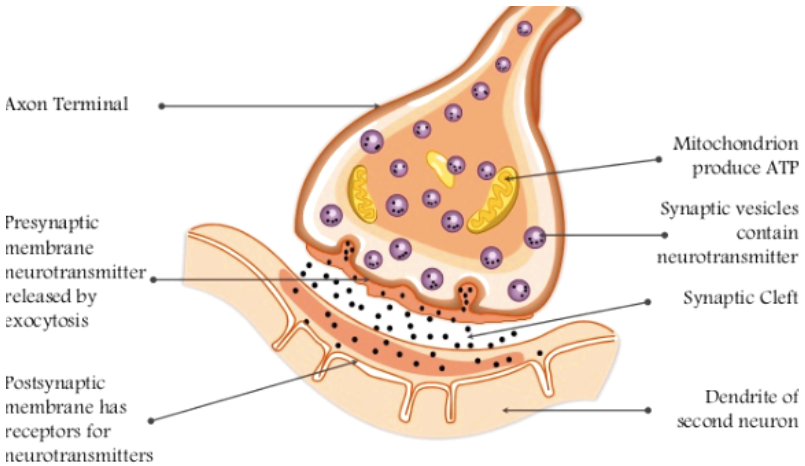
3. SODIUM CHANNELS
- Neurotransmitters in synaptic cleft bind to the receptor sites on the **postsynaptic membrane**
- Diffusion across the cleft takes approximately 0.5ms

4. **NEW ACTION POTENTIAL**
- This triggers sodium channels to open
- Causes an influx of sodium ions into the postsynaptic membrane
- Postsynaptic membrane **depolarises**
- May/may not meet the required threshold to generate an impulse in the postsynaptic neuron

5. DEACTIVATION OF NEUROTRANSMITTER
- Some neurotransmitters move away by **diffusion**
- Some **re-uptake** into the presynaptic knob
- Most are broken down by **enzymes** located on the postsynaptic membrane

Nerve agents alter neurotransmitters by kinking the signalling between our nerves, telling them to do things they normally do, but with altered frequency. Nerve agents block enzymes that stop the transmission of neurotransmitters, so the neurotransmitter keeps giving its message. An example of this is the chemical weapon, Sarin that stops the breakdown of acetylcholine.

Examples of neurotransmitters include:
- **Acetylcholine**
- **Histamine**
- **Dopamine**
- **Adrenaline/Noradrenaline**



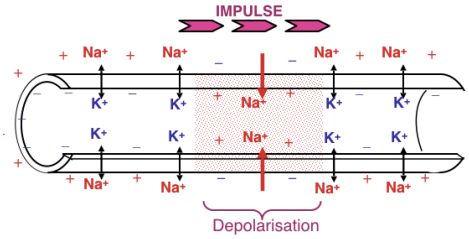
NERVE IMPULSES

The message that travels along a nerve fibre is called a nerve impulse, which is an **electrochemical change** (since it involves a change in the electrical voltage via a change in ion concentration outside the neuron)**.** They are transmitted quickly so the body is able to respond rapidly.

There are two types of charges; **positive and negative,** which repel or attract to one another.
- When opposite charges are separated, an electrical force pulls them together, releasing energy
- If separated, they have the **potential** to come together
- The potential difference between two places is known as **voltage** and is measured in **volts**
- In all body cells, there is a difference in ion concentration on either side of the cell membrane
- The potential difference created is called the **membrane potential**
- **Resting membrane potential** occurs in an unstimulated nerve cell and is 70mv less than the outside of the cell membrane

ACTION POTENTIALS

The resting membrane potential of neurons is many due to the difference in distribution of **sodium and potassium ions** on either side of the cell membrane.



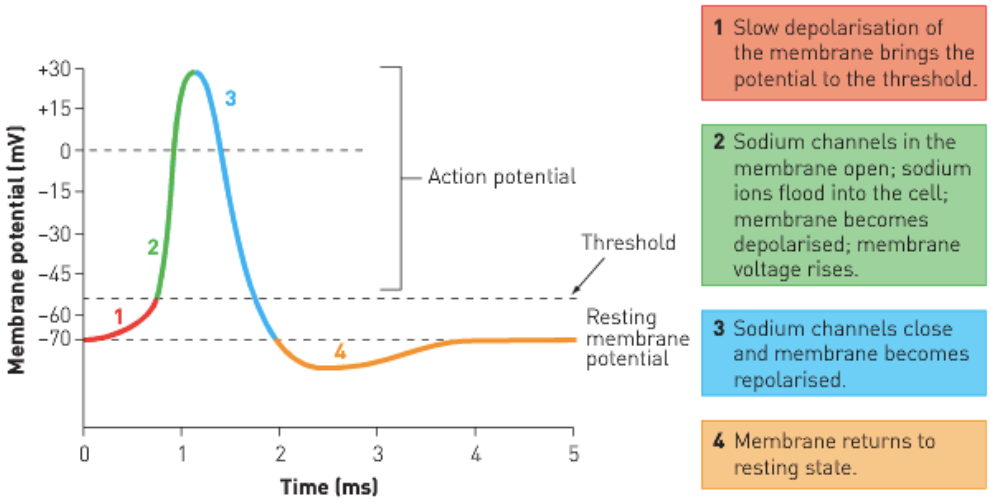
The cell membrane is:
- **Permeable** to sodium (+) and chloride (-) ions
- **Slightly permeable** to potassium (+)
- **Impermeable** to large negatively charged ions

An action potential occurs when potassium ions move out of the cell, resulting in the inside of the cell becoming negatively charged. The cell membrane maintains its potential difference in two ways:
1. Actively moves ions across the membrane via a **sodium-potassium pump** (moves sodium out of the cell and potassium into the cell)
2. Large number of negatively charged ions are trapped inside the cell, keeping it negatively charged as there is not enough potassium ions to counteract this

The membrane is **polarised** if the inside is kept negative relative to the outside of the cell.

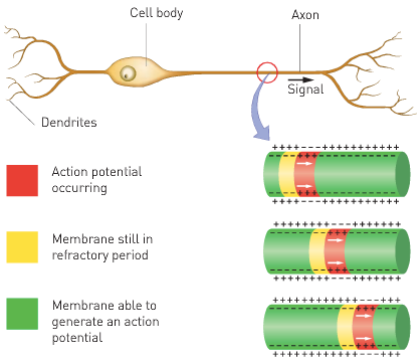
If a strong enough stimulus is applied to a nerve fibre, the membrane becomes permeable to sodium ions into the cell, which cannot be balanced by the potassium. The cell becomes **depolarised.**

- The stimulus causes a change of about 15mv
- There is an influx of sodium ions into the cell due to the sodium gated channels opening
- The cell **repolarises** as the sodium gates immediately close and the potassium gated channels open, causing the membrane to be restored to its original condition
- **All or none response**
- The **refractory period** is a brief time after the action potential where no stimulus can occur



UNMYELINATED TRANSMISSION

In an unmyelinated nerve fibre, depolarisation of one area of the membrane causes a local current flow between neighbouring areas of the membrane. This current flow causes depolarisation immediately adjacent to the site of the original stimulus.

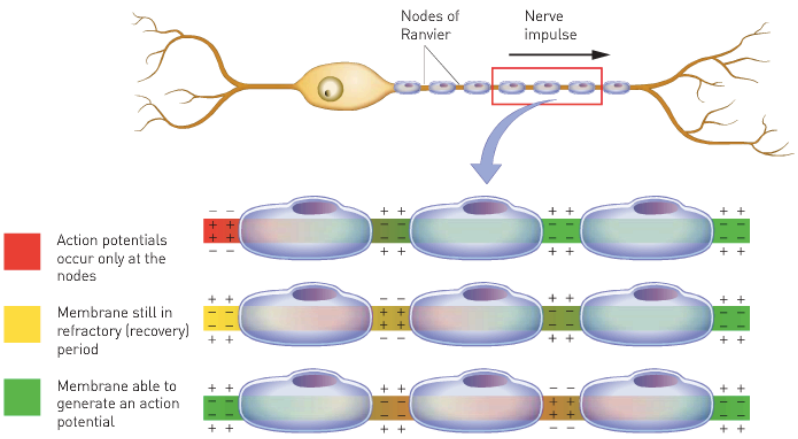


- This process repeats itself along the length of the membrane so the action potential moves away from the stimulus
- If stimulus is in middle, impulse moves both ways
- Each action potential generates another just in front of it
- Doesn't travel backwards due to the refractory period
- **Slow and steady**, with a maximum speed of 2m/s

MYELINATED TRANSMISSION

In myelinated nerve fibres, impulses travel faster as they have Nodes of Ranvier, which the impulse is able to jump across, an action called **saltatory conduction**.

- The fibres are insulated due to the myeline sheath, which stops the flow of ions outside the membrane
- Action potential therefore jumps from one Node of Ranvier to the next
- The **size of the nerve impulse is always the same**, whether it is a strong or weak stimulus
- A strong stimulus causes depolarisation of more nerve fibres than a weak one and produces more impulses at a given time



THE NERVOUS SYSTEM

DIVISIONS OF THE NERVOUS SYSTEM

The **Peripheral Nervous System (PNS)** consists of nerve fibres and a group of cell bodies called **ganglia.** Nerve fibres arise from the brain and spinal cord. There are different types:

1. **Cranial Nerves**

- 12 mixed nerves arising from the brain, carrying fibres to and from brain
- The fibres that carry impulses to the brain are **sensory fibres**
- Fibres that carry impulses away from the brain are **motor fibres**
- Cranial nerves include the optic nerve and auditory nerve

2. **Spinal Nerves**

- 31 pairs of spinal nerves arise from the spinal cord
- All are mixed nerves and are joined to the spinal cord by two roots:
    - **Ventral root** contains axons of motor neurons and have their cell bodies in the grey matter or the spinal cord
    - **Dorsal roots** contain axons and have their cell bodies in the **dorsal root ganglia**

AFFERENT AND EFFERENT DIVISIONS

Afferent division - carries impulses into the CNS by sensory neurons in the skin, joints and muscles. These nerves are **somatic sensory neurons.** It also takes impulses from the internal organs to the CNS, via **visceral sensory neurons.**

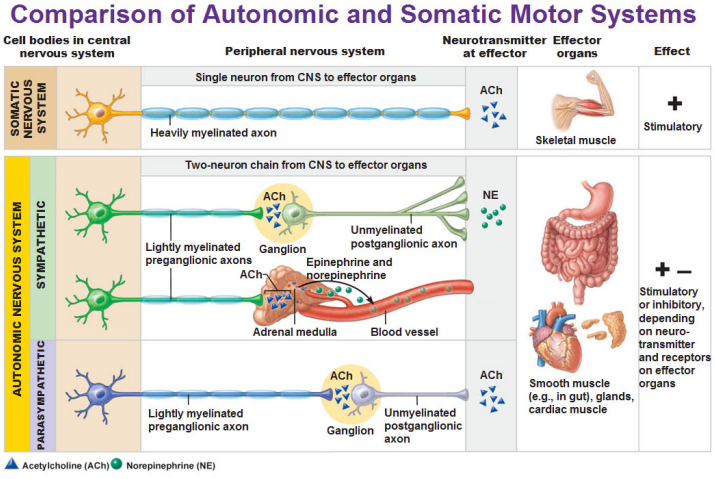
Efferent division - carries impulses away from the CNS. It is divided into:

1. **Somatic Division** takes impulses to skeletal muscles from CNS
2. **Autonomic Division** carries impulses from CNS to heart, involuntary muscles and glands. Can be subdivided into sympathetic and parasympathetic.

AUTONOMIC NERVOUS SYSTEM

Is part of the PNS and is responsible for the involuntary control of the body's internal environment. It operates via cells in the **medulla oblongata, hypothalamus and cerebral cortex.**

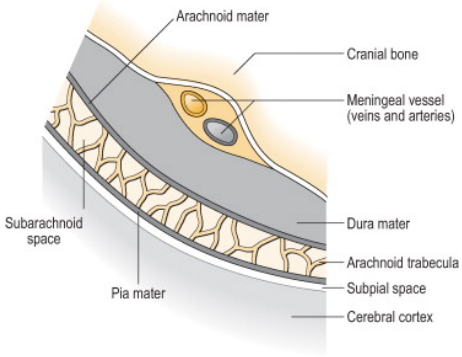
- Pathway of a impulse from the CNS to an organ is controlled by two neurons
- One has cell body in ganglia and other in CNS (spinal cord)
- **Somatic pathways** have only one motor neuron whereas autonomic has two



THE BRAIN

The brain and spinal cord are heavily protected by:

1. Bone
2. Membrane called **meninges**
3. Fluid called **cerebrospinal fluid** (CSF)



Outermost protective layer is bone called the **cranium** which the spinal cord runs through an opening in the vertebrae called the **vertebral canal**. Inside the bones are the meninges.

- The meninges cover the entire CNS
- The outer layer is called **dura matter** and is tough and fibrous, sticking close to skull
- The middle layer is called the **arachnoid matter** and is a loose mesh of fibres
- The inner layer is called the **pia matter** and has many blood vessels that give nutrients to the brain

The third protective layer is the **cerebrospinal fluid (CSF)** which occupies space between the middle and inner meninges as well as the corpus callosum. It is clear, watery fluid containing; proteins, glucose, urea, salts and cells. It has three roles:

1. Acts as a **shock absorber**
2. **Supports** the brains structure
3. **Transports** nutrients to inner cells of brain and removes wastes

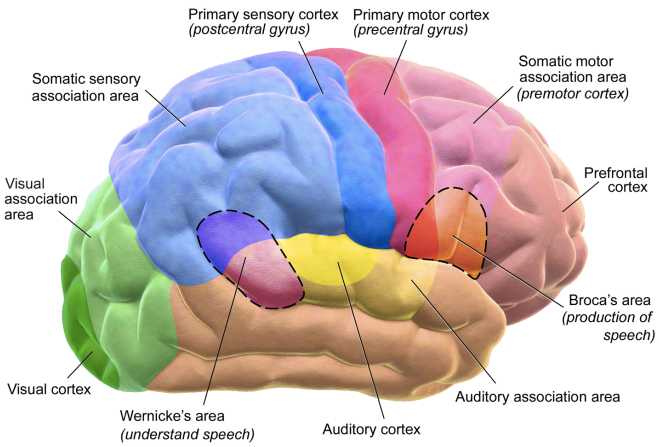
THE CEREBRUM:

Within the CNS, bundles of nerve fibres are called tracts and there are three types that occur in the white matter:

1. Tracts that contain various areas of the cortex within the same hemisphere
2. Tracts that carry impulses between hemispheres
3. Tracts that connect the cortex to other parts of the CNS

The cerebrum is involved in thinking, learning, memory, intelligence and responsibility. There are three types of functional areas in the cerebrum:

1. **Sensory areas** - interpret impulses from receptors
2. **Motor areas** - control muscular movement
3. **Association areas** - concerned with intellectual and emotional processes



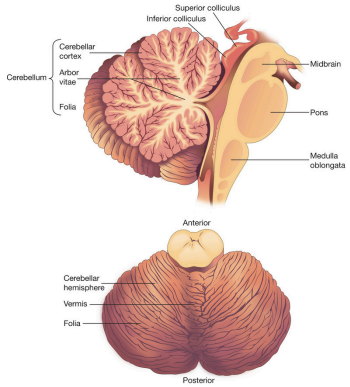
The **basal ganglia** controls skeletal muscles and memories are stores in **memory cells** in the brain.

SECTIONS OF THE BRAIN

THE CORPUS CALLOSUM

- Wide band of nerve fibres that lie underneath the cerebrum at the base of the **longitudinal fissure**
- Connects each hemisphere and allows them to communicate with each other

THE CEREBELLUM



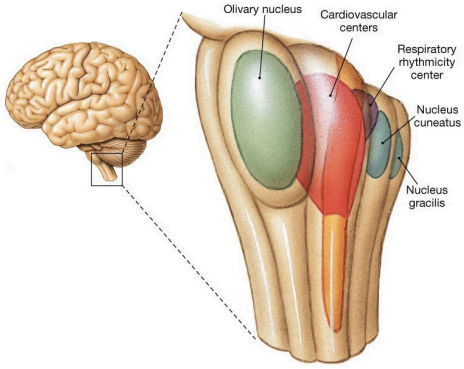
- Lies underneath the rear part of brain
- Folded into series of parallel ridges to increase surface area
- Controls **posture, balance, coordination** and receives information from inner ear and stretch receptors in skeletal muscles
- Functions take place **below conscious level**
- Impulses do not originate in cerebellum

THE HYPOTHALAMUS

Regulates autonomic nervous system, heart rate, blood pressure, secretion and movement of alimentary canal. It also regulates:

- Body temperature
- Food and water intake
- Emotional responses
- Hormones
- Sleeping patterns

MEDULLA OBLONGATA



The medulla oblongata is a continuation of the spinal cord. It contains three sections:

1. **Cardiac centre** - regulates rate and force of heartbeat
2. **Respiratory centre** - regulates rate and depth of breath
3. **Vasomotor centre** - regulates diameter of blood vessels

SPINAL CORD

- Cylindrical structure that exceeds from foreman magnum to second lumbar spine
- Outer meningeal layer contains fat and connective tissue for protection
- Composed of grey matter and white matter
- **Central canal** runs the length of the spinal cord and contain cerebrospinal fluid
- **Ascending tracts** are sensory axons and carry impulses to brain
- **Descending tracts** contain motor axons that carry impulses down from brain
- Integrates reflexes

RECEPTORS

A receptor is a structure that is able to detect change in the bodies internal and external environment. They can be individual or grouped together in a **sense organ.** They are only sensitive to one stimuli.

THERMORECEPTORS

- Respond to heat or cold found in skin
- Information is received and processed by hypothalamus and cerebrum
- Skin thermoreceptors respond to heat OR cold of external environment
- Core temperature is monitored by thermoreceptors in the hypothalamus
- Detect and regulate temperature of the blood

OSMORCEPTORS

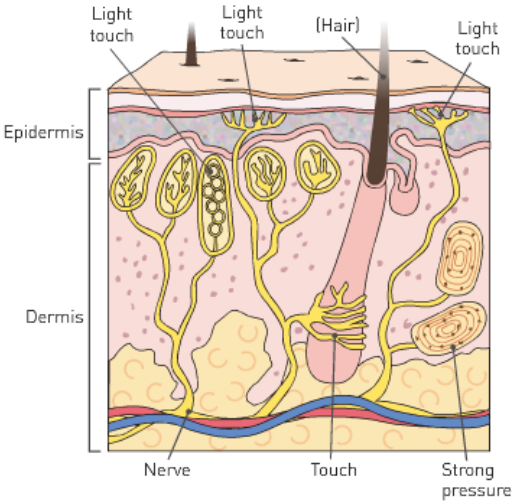
- Located in the hypothalamus and are sensitive to osmotic pressure
- Respond to very small changes in pressure and are able to stimulate hypothalamus to start water regulation

CHEMORECEPTORS

- Stimulated by particular chemicals and present in nose and mouth
- Internal chemoreceptors are sensitive to composition of body fluids
- Receptors are sensitive to pH of blood and concentration of carbon dioxide and oxygen in the blood plasma
- Involved in regulation of breathing and heart rate

TOUCH RECEPTORS

- Found in skin
- Greater concentration in sensitive areas such as the lips, fingers and eyelids
- Also associated with the base of each hair follicle which respond to light touches to the hair
- Other receptors are located deep in dermis and respond to pressure and vibrations



PAIN RECEPTORS

- Stimulated by damage to tissues, poor blood flow to tissues or by excessive stimulation
- Concentrated in skin and mucous membrane of most organs
- Adapt little or not at all so pain continues as long as a stimulus is present

REFLEXES

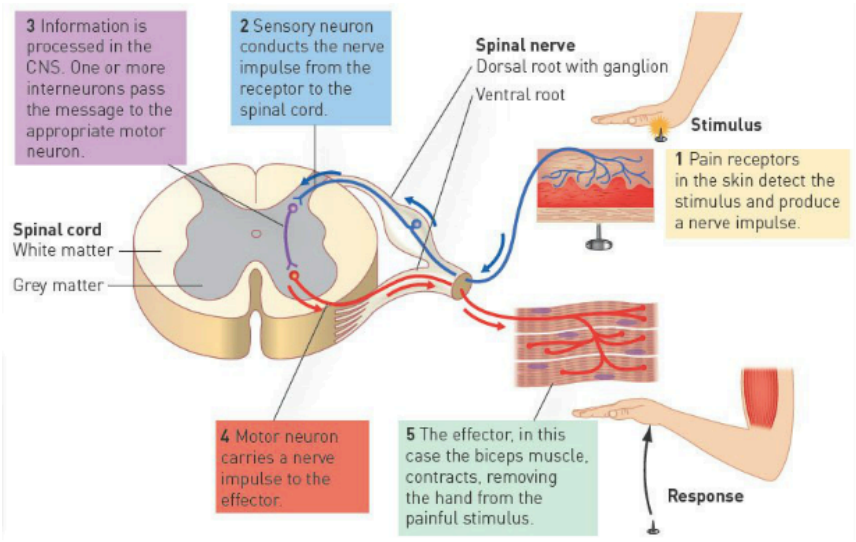
A reflex is a rapid, autonomic response to a change in the external or internal environment. It has four properties:

1. **Stimulus** is required to trigger a reflex
2. Reflex is **involuntary**
3. It is a **rapid response**
4. The response is **stereotyped**

Reflexes are usually coordinated by the spinal cord, known as a **spinal reflex.** The pathway a reflex follows from a receptor is known as a **reflex arc.**

A reflex has the following components:

1. A receptor is either the ending of a sensory neuron or specialised cell and reacts to a change by releasing an impulse
2. Sensory neuron carried impulse to CNS from receptor
3. There is at least one synapse
4. Motor neuron carries nerve impulse to an effector
5. An effector receive the nerve impulse and carries out an appropriate response.



LEARNED REFLEXES

- Reflexes such as sucking, chewing or following movement with eyes innate reflexes determined genetically
- Complex motor patterns are learned and called **acquired reflexes.**
- Learned through constant repetition
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COMPARISONS

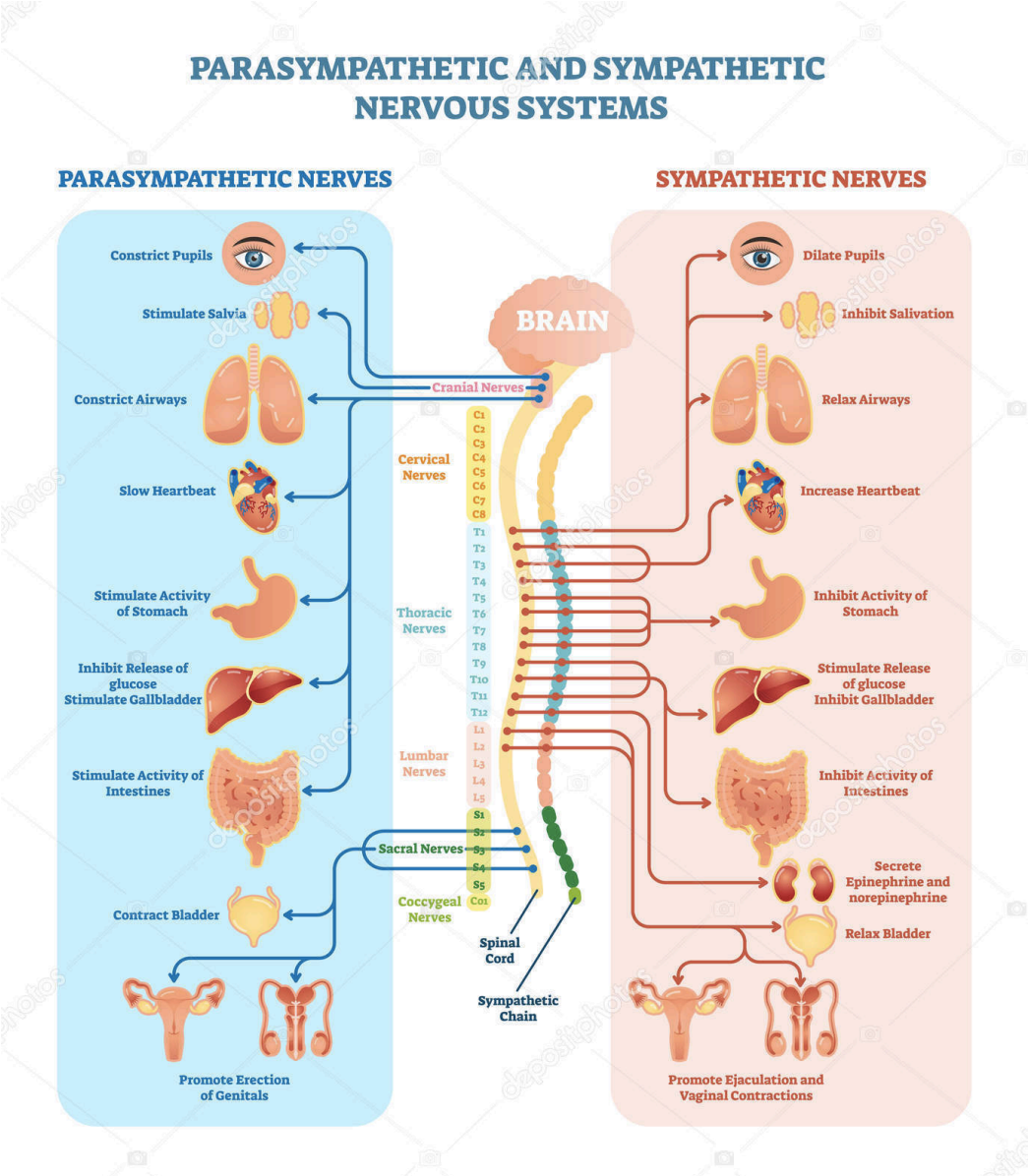
NERVOUS VS ENDOCRINE

BASIS FOR COMPARISON	NERVOUS SYSTEM	ENDOCRINE SYSTEM
The rate of response	Quick response, by the action potentials and neurotransmitters.	Responds slowly by secreting hormones, traveling through the circulatory system to the target tissue
Kind of response	Localised response.	The response is spread widely.
Duration	Short lasting effects	Long lasting effects
Transmission of signal	Neurotransmitters along neurons transmit electrochemical signals.	Hormones are chemical signals through the blood stream
Transmission	Nuerons	Bloodstream

SOMATIC VS NERVOUS

Somatic Nervous System	Autonomic Nervous System
Regulates the **voluntary** movement of the body	Regulates the **involuntary** movement of the body
Regulates movements of the body via the **skeletal muscles**, along with sensory stimuli related to vision, smell, taste, pain, noise, touch, and temperature	Regulates bodily functions such as respiratory rate, heart rate, urination, digestion, sexual arousal, pupillary response, and digestion via **glands, heart and involuntary muscles**
Made up of the **afferent nerves (sensory nerves) and efferent nerves (motor nerves)** that stimulate skeletal muscle movement	Made up of a complex network of **motor neurons**, which control glands, cardiac muscles, and smooth muscles
Divisions include the **spinal nerves** and the **cranial nerves**	Divisions include the **sympathetic and the parasympathetic** nervous system
One neuron acts as a link between the central nervous system and the effector cells	**Two neurons** act as a link between the central nervous system and the effector cells
Releases **acetylcholine** to the effector cells	Releases **acetylcholine** and **noradrenaline** to the effector cells

PARASYMPATHETIC AND SYMPATHETIC



DISEASES AND DISORDERS

NERVOUS SYSTEM DISORDERS

Two nervous system degenerative conditions that have no cure are **Parkinson's Disease and Alzheimers Disease**. Although there is no cure, **cell replacement therapy** can be used to inject cellular material into the patient and replace their dying neural tissue with healthy tissue.

Tissue engineering is the use of combination of cells, engineering materials and suitable biochemical factors to improve or replace biological functions in an effort to improve clinical procedures for the repair of damaged tissues.

- Cells are induced to grow on a scaffold which supports the cells until they can manufacture their own structure
- This is then implanted into the patient
- New tissue is then developed in the body

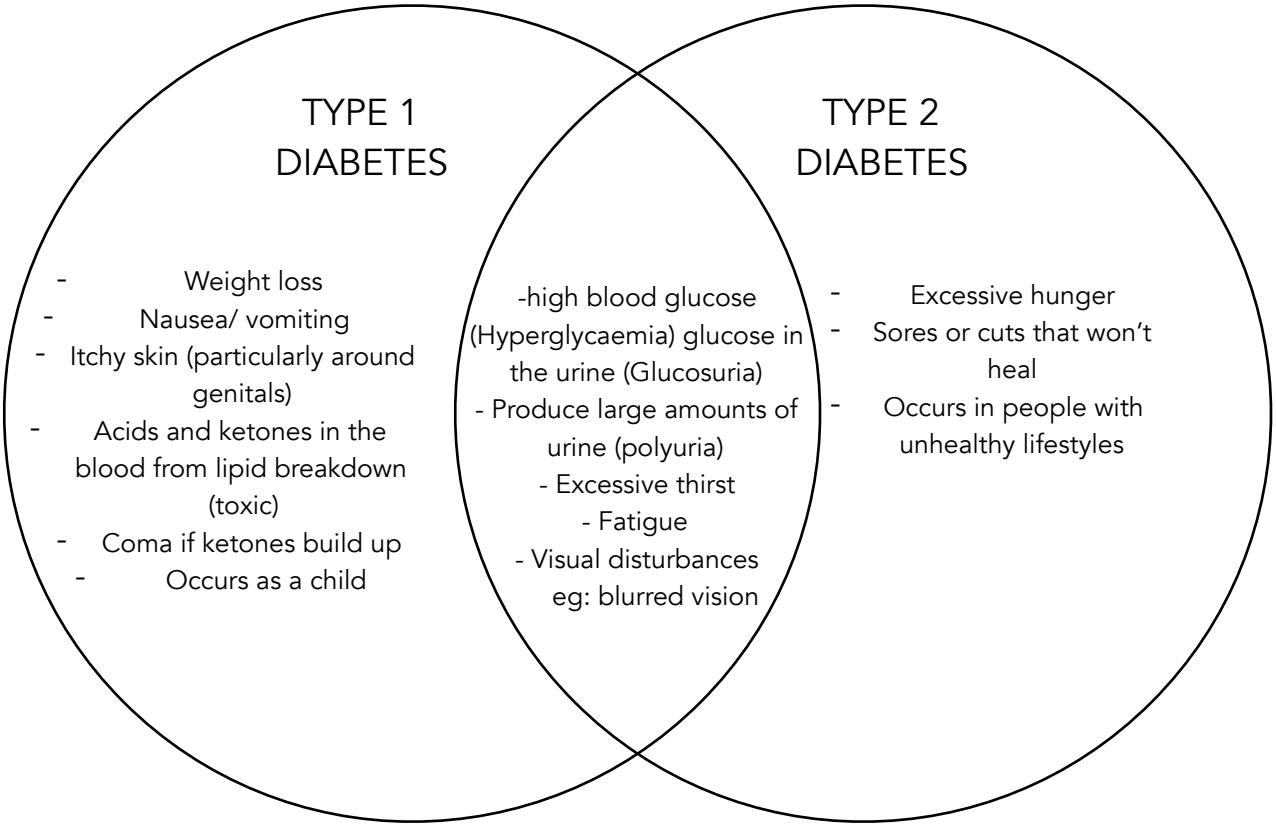
DISEASE	CAUSES	SYMPTOMS	TREATMENT	EFFECTS	PREVENTIONS
PARKINSON'S DISEASE	Reduce neurotransmitter **dopamine,** resulting in death of nerve cells	Slow physical movement and spasmodic muscle tremors	- No cure - Increasing body's dopamine levels - Cell replacement of dying neurons with healthy ones	- Impaired hearing and vision - Short term memory loss - Slow reaction times - Loss of fine motor skills	- Lower alcohol consumption - Physical stimulation - Healthy diet
ALZHEIMER'S DISEASE	Loss of neurons due to abnormal accumulations of amyloid plaques which interferes with synaptic transmission	Memory loss, paranoia, moodiness and disorientation	- No cure - cholinesterase inhibitors - Cell replacement of dying neurons with healthy ones		

ENDOCRINE SYSTEM DISORDERS

Many hormones are involved in homeostasis. Either too much or too little of these hormones will cause a disruption to homeostasis.

Diabetes Mellitus is a hormonal disorder that causes disruptions to homeostasis. It causes abnormally high blood glucose level called **hyperglycaemia.**

DISEASE	CAUSES	DIAGNOSIS	TREATMENT	PREVENTIONS
TYPE 1 DIABETES	Due to fault in immune system causing destruction of beta cells in pancreas (no insulin is produced)	Blood fasting test and finger prick test to test blood glucose levels	- No cure - Regular injections of insulin - Recombinant DNA used to insert insulin cells into body	- Genetic, no prevention
TYPE 2 DIABETES	Beta cells produce insulin but the body cells are unresponsive to it		- No cure - Medication	- Physical exercise - Healthy diet - No smoking



The over/under secretion of the **thyroid hormone**s can cause disruptions to homeostasis. The thyroid gland secretes **thyroxine** and **triiodothyronine** which both contain iodine and control the cells metabolic rate.

DISEASE	CAUSES	SYMPTOMS	TREATMENT	DIAGNOSIS	PREVENTIONS
HYPERTHYROIDISM	thyroid gland produces too much hormone thyroxine	Cells are overstimulated, rapid heartbeat, weight loss, increased appetite, fatigue, sweating, anxiety	- Drugs that block the thyroid gland's use of iodine - Surgery to remove thyroid gland	- Blood tests to test iodine, TSH and thyroxine levels in blood	- Genetic
HYPOTHYROIDISM	Insufficient amount of thyroxine produced by the thyroid gland	Metabolic processes are decreased, slow heart rate, weight gain, fatigue, lack of tolerance to the cold, swelling of the face and 'goitre'	- Iodine tablets if due to iodine deficiency - Recombinant DNA technology used to make T3 & T4		- Diet high in iodine

HUMAN BIOLOGY TOPIC 3

PATHOGENS

Communicable or infectious diseases are caused by foreign organisms invading the body and multiplying. They are contagious and spread by contact or vectors (immediate hosts of the pathogen).

BACTERIA:

- majority of bacteria are harmless to humans
- Bacteria lives on our skin, alimentary canal and other parts of the body
- All consists of one cell and their is used to classify them into species

Bacteria have many types of **structures**, their basic structure includes:

Slime layer - around outside of bacteria

Cell membrane - similar to that of other cels

Cell wall - often made of peptidoglycan, a combined carbohydrate-protein

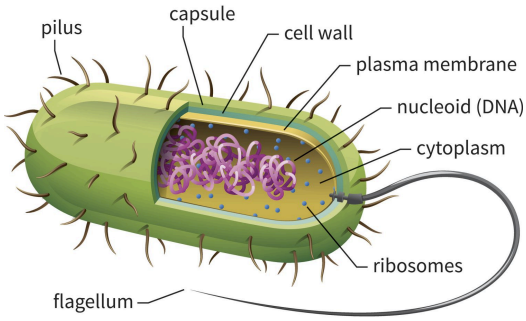
DNA - formed as a tangle inside the cell called plasmids

Flagella - for movement, but is not found in all bacteria

Cytoplasm - granular due to presence of ribosomes

No membrane-bound organelles

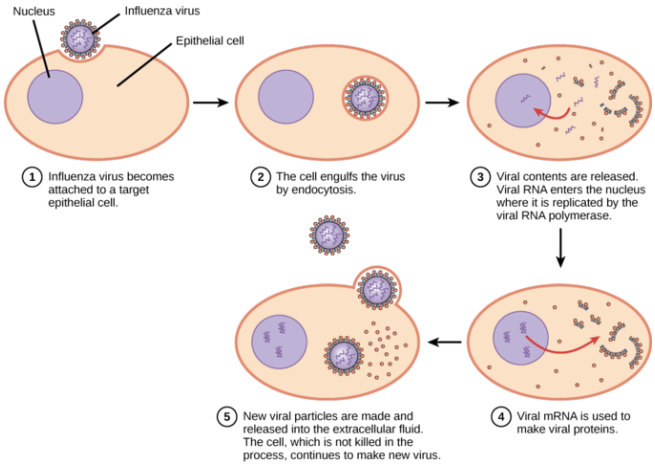
Capsule - formed of complex carbohydrate by some bacteria for protection



VIRUSES:

- too small to be seen under an ordinary light microscope
- Have a distinctive structure and differing sizes
- Have either molecule DNA or RNA, not both, surrounded by a coat of proteins
- When it infects a cell, its DNA/RNA causes cell to manufacture more of the virus
- **Bacteriophages** are viruses that attack bacterial cells

Viruses are unable to reproduce independently and therefore need a **host cell** to enable it to replicate its DNA or RNA.



	BACTERIA	VIRUSES
LIVING/NON LIVING	Living organisms, live independently	Non-living organisms as they cannot reproduce by themselves (need host)
SIZE	- Single celled - 200 to 5000nm	- 20 to 40nm in diametre - Not seen under normal light microscope
STRUCTURE	- Has a cell wall made of carbohydrate-protein (peptidoglycan) - Contains slime layer - Granular due to ribosomes - No nucleus or organelles	- True protein coat over nucleic acid - Lipid-protein envelope - No cell wall, nucleus or organelles
RNA OR DNA	- Contains both - Forms a tangle inside the cell called plasmid	Has either DNA or RNA not both
DISEASES	- Syphilis - Salmonella - Bubonic Plague - Chlamydia - Tuberculosis	- Influenza - HIV/AIDS Measles - Ebola - Chickenpox
TRANSMISSION	- Direct contact - In water droplets - Touching infected surfaces	- Person to person via body fluid - Environment to person - Animal to person
REPRODUCTION	Binary Fission	Host cells
TYPES	- Cocci (spherical) - Bacilli (Rod shaped) - Spirilla (twisted cells) - Vibrio (tear shaped)	N/A

TRANSMISSION OF DISEASES

Communicable diseases may be spread by the transmission of a pathogenic organism from one person to another. This can occur in many ways:

Transmission by contact - involves the spread of the pathogen by physical contact, either directly or indirectly. Skin infections can be spread via this method.

Transmission by body fluids - when blood or other body fluids from an infected person come into contact with the mucous membrane or blood of another person, pathogens may enter the body. Hepatitis B and HIV can spread via this method.

Infection by droplets - occur when tiny droplets of moisture, harbouring pathogenic organisms are emitted when breathing, sneezing or coughing. Measles, mumps and influenza are spread via this method.

Ingestion - of food and drink contaminated with pathogens may result in disease. Salmonella is spread via this method.

Airborne Transmission - when moisture containing pathogens evaporates, most of the bacteria is killed but bacteria and viruses remain viable and infection occurs when inhaled. The cold is spread via this method.

Transmission by vectors - transfers pathogens by other animals such as mosquitoes and ticks. Some spread it directly to the humans and others can spread it via contact with food or water. Malaria and Ross River virus are spread using this method.

PROTECTIVE REFLEXES

1. **Sneezing** - stimulated by irritation of walls of the nasal cavity, usually by fumes or dust carrying micro-organisms. Forceful expulsion of air from the lungs carries foreign particles, mucus and irritating gases out through the nose and mouth.
2. **Coughing** - stimulated by irritation to lower respiratory tract (bronchi and bronchioles). Air is forced from lungs to remove mucus and foreign particles from the body.
3. **Vomiting** - physiological stimuli, excessive stretching of the stomach from the contraction of the muscles around the abdomen, expelling the stomachs contents
4. **Diarrhoea** - irritation of the small and large intestines by bacteria. Causes quick contractions so materials pass through quickly.

NON-SPECIFIC DEFENCE

EXTERNAL DEFENCE

BODY'S EXTERNAL DEFENCE AGAINST PATHOGENS			
	LOCATION	SPECIFIC FEATURE	ACTION
SKIN	SKIN	water-proof barrier	- prevents pathogens penetrating and entering the internal system - bacterial colonies on surface make entrance of pathogens difficult
	SEBUM	oily secretion that is slightly acidic	- acidity makes environment hostile to may pathogens
	SWEAT	secretion of water, salts, wastes and fatty acids from skin	- salt and fatty acid prevents growth of pathogens
DIGESTIVE TRACTS	MUCOUS MEMBRANES	secretes mucus onto inner lining of digestive tract	- prevents bacteria entering the organs of the body
	SALIVA	contains **lysozyme**, an enzyme that kills bacteria	- creates a flushing or cleansing action that eliminates bacteria
	ACID SECRETIONS	acidity kills bacteria and reduces their growth	- creates a hostile environment for bacterias
URINOGENITAL TRACTS	URETHRA	prevents build up of pathogens	- creates a flushing or cleansing action that eliminates bacteria
RESPIRATORY SYSTEM	MUCUS	secretes mucus into nasal cavity	- traps pathogens
	CILIA AND HAIRS	tiny hairs that trap micro-organisms	- move pathogens out of respiratory tract with wave-like contractions
THE EAR AND EYE	CERUMEN	slightly acidic and contains lysozyme	- breaks down bacteria and prevents entry
	FLUSHING ACTION	contains lysozyme	- tears prevent bacteria from growing

INTERNAL DEFENCES

PHAGOCYTES

These are cells that attack organisms that penetrate through our external defences. They're Abe to engulf and digest micro-organisms and cell debris. There are two types:

Leucocytes - also known as white blood cells. They're able to lead blood capillaries and migrate through tissue to places of infection/injury. Some types secrete bacteria-killing substances whilst others engulf live bacteria and digest it.

Macrophages - large phagocytic cells that develop from some leucocytes. Some are wondering cells that look for pathogens whilst others are fixed and let pathogens find them. They either engulf and digest micro-organisms or release substances that destroy them.

PHAGOCYTOSIS:

1. Phagocyte is attracted to foreign antigen and ingests it
2. A vacuole forms inside the phagocytic cell. A lysosome binds to the vacuole and releases digestive enzymes
3. This breaks down the microbe leaving only soluble debris
4. Leaves the phagocyte by exocytosis

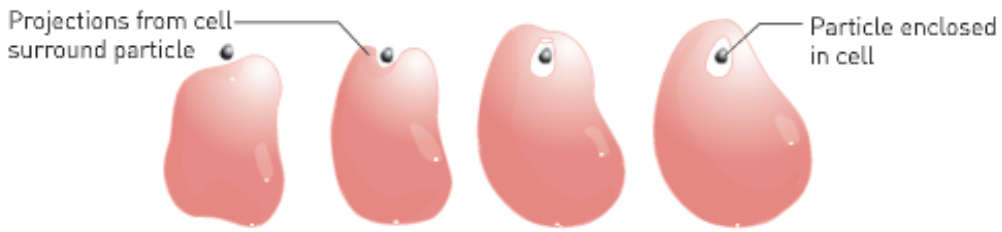


Figure 10.5 The process of phagocytosis

INFLAMMATORY RESPONSE

Inflammation is a response to any tissue damage; its purpose it to:

1. Reduce spread, destroy and prevent entry of pathogens
2. Remove damaged tissue and cell debris
3. Begin repair of damaged tissue

The four signs of inflammation are; **redness, swelling, heat and pain.**

THE INFLAMMATORY RESPONSE

1. MAST CELLS
When stimulated by mechanical damage or by local chemical damage, mast cells release histamine, heparin and other substances into the tissue fluid

NOTE: Mast Cells are cells present in most tissues that stimulate and coordinate inflammation by releasing chemicals

2. HISTAMINE
increases blood flow through the area and causes the blood vessels to become more permeable so fluid is filtered from the blood, causing heat and swelling to area of inflammation

3. HEPARIN
prevents clotting in the IMMEDIATE area of the injury and instead clots form AROUND the injury, stopping the speed of pathogens to healthy tissues

4. PHAGOCYTES
the chemicals released by mast cells attract phagocytes; macrophages and leucocytes, that consume micro-organisms and debris by phagocytosis

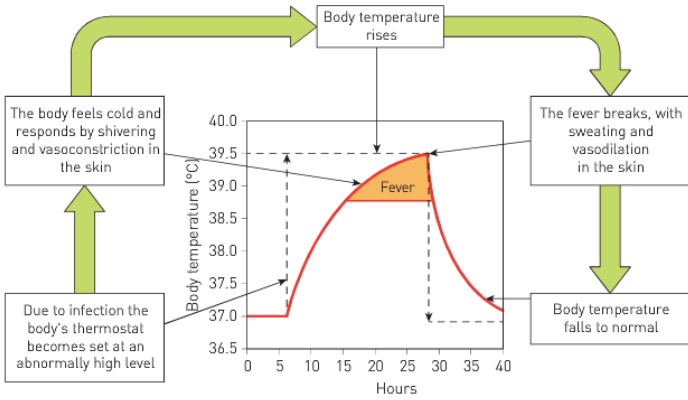
5. PAIN RECEPTORS
stimulated so pain is felt in inflamed area

6. PUS AND NEW CELLS
Phagocyte death forms a pus and new cells are produced by mitosis to repair damaged tissue

FEVERS

During an infection, there is an elevation in body temperature called a **fever**. This is due to the resetting of the **body's thermostat** to a higher level, which is controlled by the hypothalamus.

- When experiencing rapid fevers, the person feels cold and vasoconstriction/shivering occur
- Increases and conserves heat
- Once body temperature reaches a point called a **crisis**, the fever breaks and sweating/vasodilation occur
- Fever **inhibits the growth of some bacterias and viruses**
- Release of **pyrogens** by white blood cells reset the body's thermostat
- Temperature over 44.4 degrees is deadly to individual



LYMPHATIC SYSTEM:

The lymphatic system consists of:

1. A network of lymph capillaries joined to larger lymph vessels
2. Lymph nodes located along the length of the vessels

The main function is to **collect fluid from blood capillaries** and return it back to the circulatory system. It is also important part of the body's internal defence system against pathogens.

- Lymph entering lymph nodes contain cell debris, foreign particles and micro-organisms that have penetrated the body's defence
- Can cause disease if not destroyed
- Lymph nodes contain masses of l**ymphoid tissue** which traps bacteria and large particles
- Macrophages destroy the particles by phagocytosis
- Lymph nodes become swollen and sore

GOOD HYGIENE PRACTISES

- **Cover mouth** when coughing or sneezing to reduce spread of bacteria
- **Wash hands** thoroughly when eating, using the toilet, cleaning etc
- **Wear gloves** when cleaning up blood or body fluids
- Use **mechanical barriers** such as a surgical mask or condoms in certain situations

SPECIFIC DEFENCES

Specific defences are directed towards a particular pathogen, for example if you contract chicken pox, your body will make specific antibodies to combat this disease. Specific defences are part of the **immune system.**

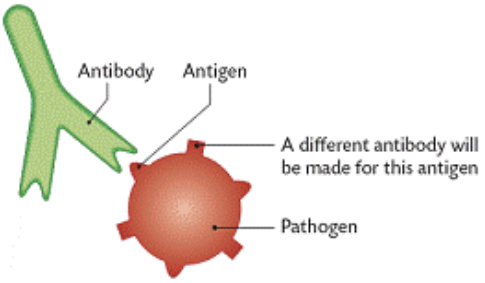
THE IMMUNE RESPONSE:

This is a **homeostatic** mechanism that helps deal with the invasion of pathogens and restore the body's internal environment.

- Lymphoid tissues are found in lymph nodes, spleen, thymus and tonsils
- They are composed of **b-lymphocyte**s (B cells) and **t-lymphocytes** (T cells)
- B cells provide antibody mediated immunity and T cells provide cell mediated immunity
- Both are produced in the bone marrow
- T cells matures in the **thymus** before incorporated into the lymphoid tissue
- B cells mature in the **bone marrow** before part of the lymphoid tissue

ANTIGENS:

All immune responses are triggered by **antigens**. An antigen is any substance cable of causing a specific immune response causing the body to produce **specific antibodies.**



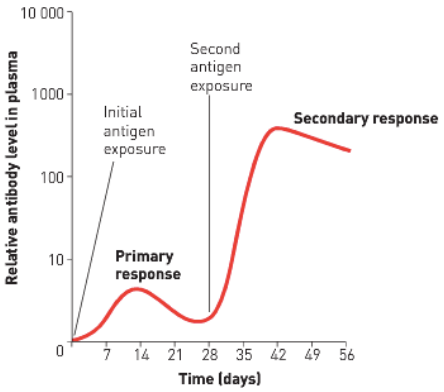
- Are large molecules composed of proteins, carbohydrates, lipids or nucleic acid
- Antigens are found on the surface of all cells
- Molecules produced by the individual are **self-antigens** or **non-foreign antigens**
- The body does not attack these unless it has an autoimmune disorder
- Foreign compounds that trigger an immune response are **non-self antigens** or **foreign antigens** and the body attacks these

ANTIBODIES:

Antibodies are specialised **globular proteins** produced in response to a non-self antigen. They belong to a group of proteins called **immunoglobulins** (Ig), secreted by B lymphocytes.

- They combine with antigens to create an **antigen-antibody complex**
- Have specific active sites where they combine with an antibody
- Immobilise foreign cells via **agglutination**

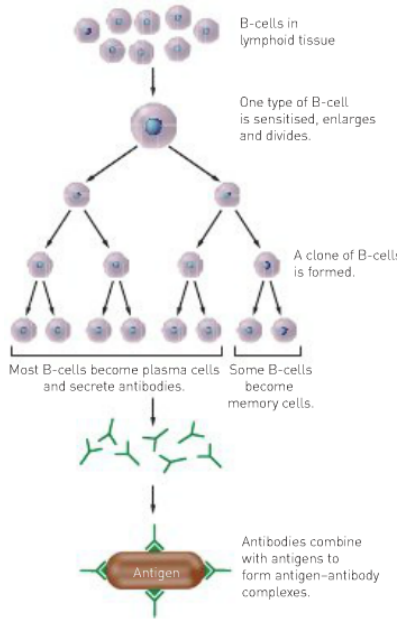
ANTIBODY MEDIATED RESPONSE



Is also known as **humoral response,** it involve the production and release of antibodies into the blood and lymph, providing a resistance to viruses, bacteria and bacterial toxins before they enter the body's cells.

PRIMARY RESPONSE:

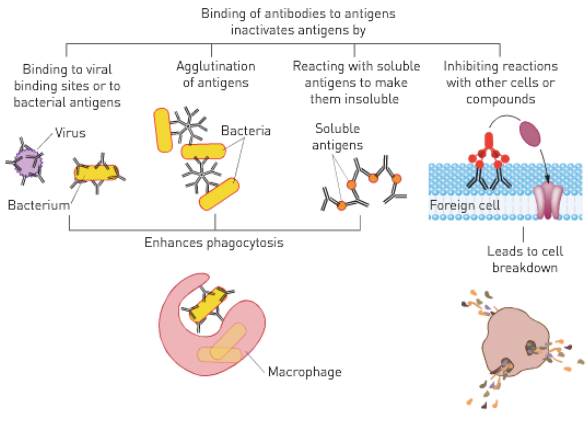
- First exposure to an antigen produces a slow response that takes days for cells to divide and differentiate
- When the B cells in the body are stimulated by a foreign substance, they enlarge and divide by mitosis to form a group called a **clone**
- Sensitised B cells are produced and mature in bone marrow
- Most of these become **plasma cells** and some are **memory cells**
- Plasma cells secrete specific antibodies that attach to the active site on the antigens
- Released into bloodstream to find site of invasion
- Remaining memory cells build a resistance to the infection if it was to attack again



SECONDARY RESPONSE:

- much faster as memory cells help plasma cells more quickly
- Antibody levels in blood rise more rapidly
- Antibodies work by combining with antigens to create **antigen-antibody complex**
- They inactivate and destroy non-antigens by:

1. **Binding** to viral binding sites or to bacterial antigens
2. **Agglutination** (clumping non-self particles together)
3. Reacting with soluble antigens and making them **insoluble**
4. **Coat bacteria** to enhance phagocytosis
5. **Inhibit reactions** with other cells or compounds by breakdown of non-self cell



CELL MEDIATED IMMUNITY

Cell-mediated or **cellular immunity** provides resistance to the intra-cellular phase of bacterial and viral infections. They specialise in invading and replicating inside their hosts own cells, making them difficult to overcome. Also important in the rejection of transplants, foreign tissue and fighting cancer cells.

- T cells are sensitised and undergo rapid cell division
- Only occurs after B cells or macrophages encounter the antigen and present it to the T cells
- The T cells become four different types of cells:

1. Killer T Cells

Migrate to the site of infection and deal with invading antigens by attaching to the invaders and secrete a substance that destroys them

2. Helper T cells

Secrete a number of substances that causes lymphocytes at infection sites to become sensitised and therefore intensifying their response, attract macrophages to destroy the antigens by phagocytosis and intensify the phagocytic activity of macrophages

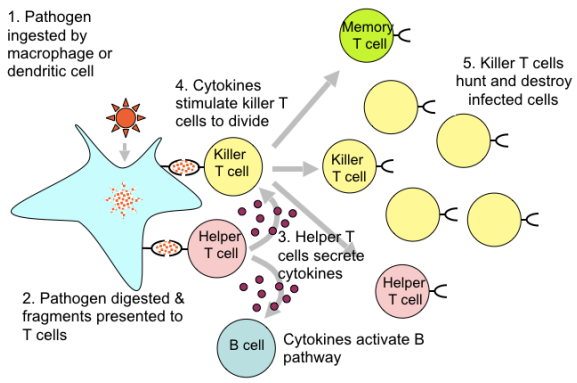
3. Suppressor T Cells

Act when response has been successful and release substances that inhibit B cell and T cell activity, slowing the immune response.

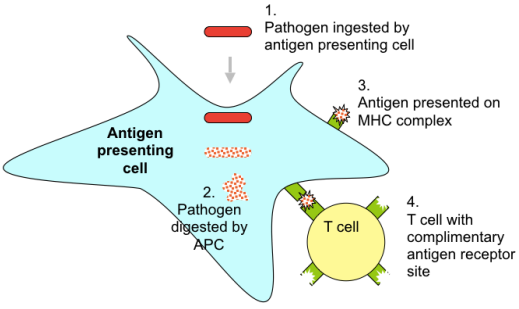
4. Memory Cells

Hold information in producing plasma cells if antigen was to invade again.

CELL MEDIATED



ANTIBODY MEDIATED



IMMUNITY

Immunity is the resistance to infection by invading micro-organisms. The body's ability to respond quickly to pathogens may be **natural** (without human intervention) or **artificial** (given an antibody or antigen).

- **Passive immunity** is when a person is given antibodies produced outside their own bodies
- Naturally happens from mother to foetus via placenta and breastmilk
- Gain artificially through injections
- **Active immunity** is when the body is exposed to foreign antigens and manufactures its own antibodies in response
- Occurs due to infection or an injection of antigens

	NATURAL (occurs without human intervention)	ARTIFICIAL (results from people being given either an antigen or antibody)
PASSIVE	- person is given antibodies from someone else - immune system NOT activated - NO memory cells formed (= NO immunity acquired) - protection immediate but only temporary - Not long lasting ~ 2 weeks - Eg: mothers breast milk/ placenta passes antibodies to foetus	- person is given antibodies from someone else - immune system NOT activated - NO memory cells formed (= NO immunity acquired) - protection immediate but only temporary - (NOT long lasting) - Eg: influenza/tetanus/rabies antibodies injected into bloodstream to combat an infection
ACTIVE	- Immune system IS activated - Body makes own antibodies in response to foreign antigen - memory cells ARE created - Long lasting immunity - Eg: chicken pox	- Person is given antigens - Body makes own antibodies in response to foreign antigen - memory cells ARE created - Long lasting immunity - Eg: antigens given in vaccination: living attenuated MMR injected into bloodstream

VACCINATIONS

Immunisations and **vaccines** are programming the immune system so that the body can respond rapidly to infecting micr-organisms. They allow appropriate antibodies to be produced without the person actually having to suffer that disease.

Herd immunity is when a large proportion of a community is immunised, meaning there is a lower chance of the pathogen spreading.

TYPE OF VACCINE	EXPLANATION AND EXAMPLE
1. LIVING ATTENUATED MICRO-ORGANISMS	- Reduced virulence (less ability to infect/produce disease) - Person does NOT contract disease - Can be made by recombinant DNA technology - Eg: MMR, rabies, tuberculosis, yellow fever, poliomyelitis
2. DEAD MICRO-ORGANISMS	- Microbe killed before injected - Not as long lasting but still produces immune response - Eg: whooping cough, cholera, typhoid, bubonic plague,
3. TOXOIDS	- Toxins produced by bacteria are inactivated - Doesn't make the person ill - Eg: diphtheria, tetanus
4. SUB-UNIT	- A fragment of micro-organism is used to provoke an immune response - Eg: HPV (Gardisil), Hepatitis B

RISK FACTORS:

- Allergic reactions may occur due to the vaccine
- Cross species disease introduction
- Possible side effects which maybe be dangerous
- Longevity of protection (booster shots needed)

ETHICAL CONCERNS:

- Exploitation of those in developing countries who are not aware of risks involved
- Human foetuses that have been aborted have their cells used for Rubella vaccine
- Viruses can only be reproduced in living cells so viral vaccines require host tissues, eg influenza virus is cultured in chicken embryos
- Animal ethical concerns

ANTIBIOTICS AND ANTIVIRALS

Antibiotics are drugs used to fight infections caused by bacteria. Some are naturally occurring but most are synthetically manufactured.

The first antibiotic was **Penicillin**, derived from fungal mould. Since then, other antibiotic substances were discovered such as **streptomycin** which is made from an **actinomycete** bacteria that lives in the soil and produce branching filaments which can disrupt protein synthesis in cells of target bacteria.

Narrow-spectrum antibiotics are only effective to a specific or small range or bacteria whereas **broad-spectrum antibiotics** are effective in inhibiting growth of many different bacteria.

There are two types of antibiotics:

1. **Bactericidal antibiotics** - which kill bacteria directly by changing the structure of the cell wall or cell membrane, or disrupting the action of essential enzymes
2. **Bacteriostatic antibiotics** - prevent bacterial cells from growing, usually by preventing protein synthesis

The overuse of antibiotics has lead to bacteria becoming resistant to them. They can either be resistant to many types of antibiotics **(multiple drug resistance)** or all types of antibiotics **(total drug resistance).**

ANTIVIRALS:

Antiviral drugs are used specifically for treating **viral infections** such as influenza and HIV.

- Viruses enter the host cell and virus DNA/RNA indices the cell to produce new virus particles
- Can then affect new host cells
- Antivirals inhibit the development of the virus
- Most antivirals are targeted at HIV, Hepatitis B and C and Influenza A and B
- Now scientists are able to understand genetic sequence of viruses, more antivirals will become available in the future

HUMAN BIO TOPIC 4

VARIATION IN A POPULATION

A population is a group of organisms of the same species that live in the same place at the same time. A **gene pool** is the sum of all the alleles in a given population.

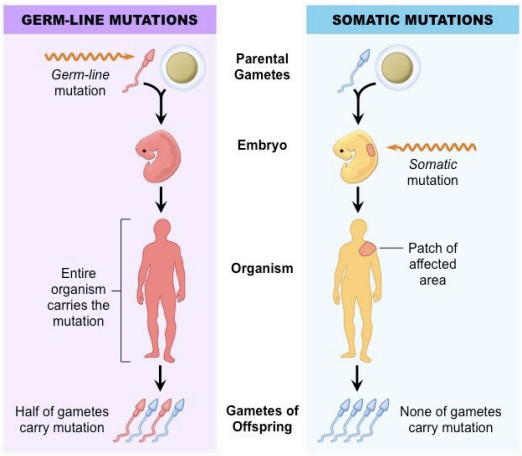
Allele frequencies in a gene pool are affected by:
1. Reproduction rates
2. Mutations
3. Migration in and out of a population
4. Natural events such as change in environment

Changes in allele frequencies allows populations to be compared at different times or in different locations.

Genetic mutation introduces new alleles and therefore new variations into a population. They occur by chance and can either be beneficial, harmful or neutral. There are two main types: **gene mutation** (change in a single gene) and **chromosomal mutations** (change in a chromosome).

MUTAGENS
- Occur without a known cause
- **Mutagenic agents** increase the rate of mutation
- Examples include: x-rays, ultraviolet light, mustard gas and bacterias

SOMATIC AND GERM-LINE MUTATIONS
- Mutations can occur in the reproductive cells of a person, called **germ-line mutations**
- If the reproductive cells are affected, the mutation can occur in the gametes and be passed on to other generations
- Example: PKU
- **Somatic mutations** only affect body cells and aren't genetic



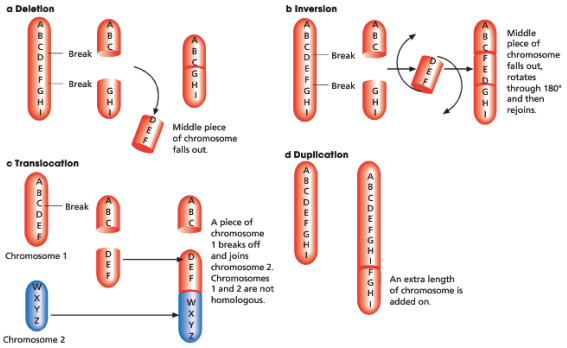
GENE MUTATIONS
Gene mutations occur during DNA replication which is then copied over and over as the cell divides. The simplest form of mutation is point mutation in which only a single nucleotide is affected.
- **Synonymous** (Silent): change in nucleotide codes for same amino acid, therefore no effect
- **Missense**: substation of a nucleotide results in a different amino acid
- **Nonsense**: occurs when substitution creates a new 'stop' codon

Albinism - due to missing pigment from hair, skin, and eye from a missing protein
Duchenne Muscular Dystrophy - arise through male zygote so child develops disease, resulting in wasting of arms, legs, shoulders and chest and causes failure of respiratory system and death
Cystic Fibrosis - caused by gene mutation on chromosome 7 which codes for protein that regulates passage of chloride ions across cell membrane, symptoms include salty skin, coughing and pneumonia

CHROMOSOMAL MUTATIONS

Chromosomal mutations involve all parts of the chromosome, affecting a number of genes. There are five main types:

1. **Deletion** - double strand breaks may cause sections of chromosomes to drop out, removing all genes. The two ends rejoin to make shorter chromosones which can be fatal to the organism
2. **Inversions** - chromosome breaks and flips 180 degrees before it rejoins, reversing the normal sequence of genes
3. **Translocation** - a section of the chromosome breaks off and reattaches with another which can result in the formation of cancers
4. **Duplication** - occurs when an extra copy of a DNA sequence is made and inserted into the chromosome
5. **Non-disjunction** - during meiosis, chromosome pairs don't divide evenly and one daughter cell has more whilst the other has less



TRISOMY

Trisomy is the addition of an extra chromosome into a daughter cell which then becomes a zygote.

1. **Down syndrome** - (trisomy 21) occurs when a when a child has an extra chromosome 21. Many of the symptoms of down syndrome may occur when partial trisomy occurs (part of C21 attaches to another chromosome)
2. **Patau Syndrome** - when an extra chromosome 13 produces individuals with mental retardation, small head, cleft lips and extra digit on each hand.
3. **Trisomy 16** - most common type of trisomy but also results in spontaneous miscarriages
4. **Klinefelter's Syndrome** - trisomy XXY produces normal boys but develop into mentally retarded adults with small testes, large breasts and missing secondary male sex characteristics

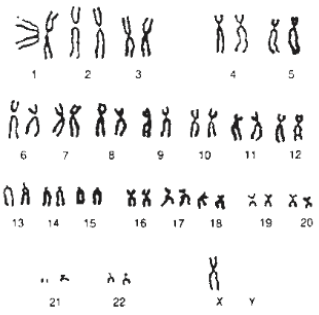


Figure 12.7 Karyotype for Turner's syndrome

MONOSOMY

Monosomy is when an individual is missing a chromosome, only having one copy instead of two. In some cases this can result in a miscarriage.

1. Cri du chat Syndrome- only part of the chromosome 5 is missing, the child has a cry sound like a cat due to problems with nervous system and larynx

2. Turner's Syndrome - monosomy X, these females are short in stature, lack secondary sex characteristics and are infertile

GENETIC VARIATION

Variation between individuals of a species are caused by a number of processes, all of which contribute to the inherited characteristics of the individual.

1. **Random assortment** - during meiosis results in games with large number of combination of chromosomes from the parents
2. **Crossing over** - of chromatids during meiosis may result in pieces of chromatid being broken off and attaching to a different chromatid, resulting in a change of sequence
3. **Non-disjunction** - where one or more pairs of chromosomes fail to separate during meiosis
4. **Random fertilisation** - the fact any sperm can fertilise any egg due to chance and each person will produce a huge number of different sperm and egg in respect to the alleles each contain
5. **Mutations** - a permanent structural alteration in an organism's DNA and may result in new characteristics of an individual

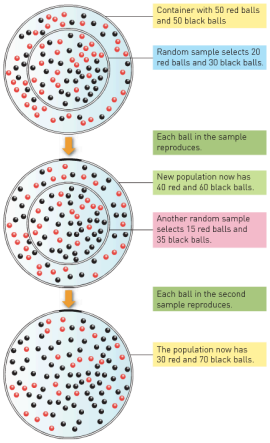
ALLELE FREQUNCIES IN A POPULATION

The proportion of alleles occurring in one generation to the next in a large population is very much the same. If **natural selection** is operating on some characteristics at the expense of others, overtime the allele frequency for this characteristic would change.

RANDOM GENETIC DRIFT

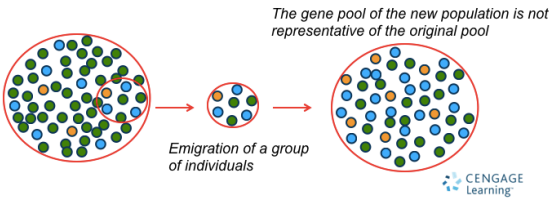
In small populations there is random, non-directional variation, known as genetic drift, which is purely a chance occurrence. It affects smaller populations where changes in allele frequencies are more noticeable and % change is more drastic

- Example: The Islands of Bentinck and Mornington were once connected to the mainland but rising sea levels cut them off = isolated their populations
- Islander populations had allele frequencies HIGH in I^B blood types, I^A was absent.
- Mainland population had allele frequencies low in I^B blood types, I^A occurred in higher frequency



Founder's Effect - when a small group moves away from the original population, founding a new area. The migrant group is small and usually their alleles do not express all of the alleles of the original population

- Example: Amish people who have moved to remote areas, at least one of the 200 had the recessive allele for Ellis-Van Creveld syndrome, making it more common amongst this small population as they interbreed



Bottleneck Effect - occurs when there is a disaster of some sort that reduces a population to a small handful, which rarely represents the actual genetic makeup of the initial population.

MIGRATION

The gene flow, or movement of genes, from one population to another. Gene flow may occur if migrants breed with one another

- Immigrants may add new alleles to one gene pool and immigrants may remove some alleles from another
- Example is the increase in B blood group in Indigenous Australians due to migration of people from Asia and Europe

BARRIERS TO GENE FLOW

Populations are separated from each other and different selective environmental pressures exist, some traits are favourable over others. Separate gene pools develop and allele frequencies show differences between the separate populations

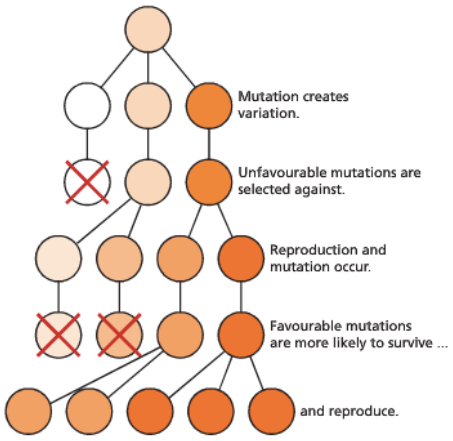
Geographical barriers - created by physical isolation and boundaries such as oceans, mountains, lake systems, deserts or ice

Sociocultural barriers - created by religion, culture, education, ethnicity and status which may prevent some people from breeding and cause inbreeding in other populations

NATURAL SELECTION

Natural selection occurs when selection pressures in the environment confer a selective advantage on a specific phenotype to enhance its survival and reproduction; this results in changes in allele frequency in the gene pool of a population over time. The principal of natural selection resulting in evolutionary change rests on the following propositions:

1. There is variation of characteristics within a species
2. More offspring are produced then what can survive to maturity
3. Because of excessive birth rate and limited resources their is a struggle for existence
4. Individuals with characteristics best suited to their environment have an advantage (survival of the fittest)
5. Favourable characteristics are passed on to the next generation
6. The allele frequency for these favourable characteristics will increase in the population



SPECIATION

Occurs when a single population becomes two seperate populations that are unable to interbreed due to changes that produce physical, biological or behavioural barriers. There are three broad processes that work together in the evolution of this great diversity:

1. **Variation** - within the population that share a common gene pool
2. **Isolation** - a barrier has formed, dividing the population in two. No interbreeding occurs and they have their own separate gene pools
3. **Selection** - different selection pressures act on the two populations over many generations, bringing a change in the gene frequencies
4. **Speciation** - over a long period of time the two groups are no longer able to interbreed to produce fertile offspring, therefore a new species is formed

TAY SACHS DISEASE

WHAT IS THE DISEASE AND IT'S CAUSES?

Tay Sachs is a fatal genetic disorder that results in progressive destruction of the nervous system. Tay-Sachs disease results from defects in a gene on chromosome 15 that codes for production of the enzyme Hex-A. Its absence causes a fatty substance, or lipid, to accumulate abnormally in cells, especially in the nerve cells of the brain. This ongoing accumulation causes progressive damage to the nervous system, resulting in death at a young age, usually before they are four or five years of age. It is inherited autosomal recessively.

WHAT POPULATION IS AFFECTED BY THIS DISEASE?

Tay Sachs is an autosomal recessive disorder that mainly affects young people of Jewish decent from Eastern Europe (Ashkenazi Jewish population), it also affects the Cajun population of Southern Louisiana due to the migration of Jewish people into this isolated population.

WHAT ARE THE MOST COMMON SYMPTOMS OF THIS DISEASE?

- seizures
- vision and hearing loss
- intellectual disability
- paralysis
- an eye abnormality called a cherry-red spot
- infants lose motor skills such as turning over, sitting, and crawling
- sensitivities to loud noises

ARE THERE ANY TREATMENTS FOR THIS DISEASE?

There is no cure or effective treatment. Scientists are exploring enzyme replacement therapy to provide the Hex-A that is lacking in babies with Tay-Sachs.

HOW HAS THIS DISEASE PERSISTED IN THE POPULATION?

People who are carriers of TSD have a heterozygous advantage as they have a resistance to tuberculosis, therefore keeping their alleles more frequent in the gene pool. People with two recessive alleles for TSD are likely to die before reproductive age, therefore not passing on their alleles, however, people with two normal alleles have less of a survival advantage if they were to contract tuberculosis.

WHAT EVOLUTIONARY PHENOMENAS ARE IN PLAY?

Genetic drift - as Jewish population are small and isolated due to discrimination
Sociocultural Barriers - due to discrimination the Ashkenazi Jewish population has been isolated into overcrowded ghettos where tuberculosis was prevalent, also shows marriage between those of same religion
Migration - Jewish people migrated into isolated Cajun society, introducing the allele for Tay-Sachs Disease, which therefore changes the frequency of the TSD allele for this population
Natural selection - the heterozygotes have a resistance to tuberculosis

HOW DOES NATURAL SELECTION RESULT IN NEW ALLELE FREQUENCIES?

1. There is variation within a population, the allele for TSD is present as well as the normal allele which causes three outcomes in a population: TSD, normal or carrier.
2. More offspring are produced than can survive to maturity, causing a struggle for existence. Those with alleles best suited to their environment are more likely to survive to reproductive age and pass on their alleles.
3. The mutation of TSD allele increases in variation within the population. Having one of these alleles has created a resistance to tuberculosis, therefore populations with a high tuberculosis frequency will also have a high number of people that are TSD carriers. The heterozygotes in this population is advantageous because it has a resistance to tuberculosis and they are not affected by the harsh affects of TSD.
4. Individuals that carry TSD will produce offspring who are likely to inherit either of the genes, therefore passing it onto their children. Those who have TSD don't survive to reproductive age and therefore cannot pass on their alleles
5. The TSD allele would eventually become more dominant gene in the population

SICKLE CELL ANAEMIA

WHAT IS THE DISEASE AND IT'S CAUSES?

Sickle cell anaemia is an inherited form of anaemia — a condition in which there aren't enough healthy red blood cells to carry adequate oxygen throughout your body. Normally, your red blood cells are flexible and round, moving easily through your blood vessels. In sickle cell anaemia, the red blood cells become rigid and sticky and are shaped like sickles or crescent moons. These irregularly shaped cells can get stuck in small blood vessels, which can slow or block blood flow and oxygen to parts of the body. Sickle cell anaemia is formed by a mutation on the gene responsible for the production of normal haemoglobin. The mutant allele is due to the substitution of the amino acid valine for glutamic acid. Sickle cell is usually referred to as haemoglobin S and therefore is shown as HbS. It is usually a fatal disease.

WHAT POPULATION IS AFFECTED BY THIS DISEASE?

Sickle cell anaemia is more common in those who have black African descent. In the tropical zone of Africa, 40% of the population of some tribes carry the sickle cell trait. Sickle cell disease occurs more often among people from parts of the world where malaria is or was common.

WHAT ARE THE MOST COMMON SYMPTOMS OF THIS DISEASE?

Anaemia - Sickle cells break apart easily and die, leaving you without enough red blood cells. Without enough red blood cells, your body can't get the oxygen it needs to feel energised, causing fatigue.
Episodes of pain - Periodic episodes of pain, called crises, are a major symptom of sickle cell anaemia. Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to your chest, abdomen and joints.
Painful swelling of hands and feet - The swelling is caused by sickle-shaped red blood cells blocking blood flow to the hands and feet.
Frequent infections - Sickle cells can damage the spleen, leaving you more vulnerable to infections.
Delayed growth - Red blood cells provide your body with the oxygen and nutrients you need for growth. A shortage of healthy red blood cells can slow growth in infants and children and delay puberty in teenagers.

ARE THERE ANY TREATMENTS FOR THIS DISEASE?

There's no cure for most people with sickle cell anaemia, but treatments can relieve pain and help prevent problems associated with the disease. Bone marrow transplant, also known as stem cell transplant, offers the only potential cure for sickle cell anaemia.

HOW HAS THIS DISEASE PERSISTED IN THE POPULATION?

A person with the sickle cell trait has a mixture of normal and faulty haemoglobin in their red blood cells without having sickle cell disease. People with sickle cell trait have enough normal haemoglobin in their red blood cells to prevent the cells from sickling. However, people with sickle cell trait can genetically pass the trait to their children. If two people with sickle cell trait have children together, there is a 1 in 4 chance that their children will have sickle cell anaemia. Sickle cell anaemia is more fatal as there is less oxygen delivered to organs as there is a higher number of sickled red blood cells.

WHAT EVOLUTIONARY PHENOMENAS ARE IN PLAY?

Natural Selection - those who are heterozygotes for sickle cell have a survival advantage over those who are homozygous for sickle cell or those who have the normal allele in areas prone to malaria

HOW DOES NATURAL SELECTION RESULT IN NEW ALLELE FREQUENCIES?

1. There is variation within a population, the allele for sickle cell is present as well as the normal allele which causes three outcomes in a population: sickle cell anaemia, normal or sickle cell trait.
2. More offspring are produced than can survive to maturity, causing a struggle for existence. Due to selection pressures, in this case malaria, alleles best suited to their environment are more likely to survive to reproductive age and pass on their alleles.
3. The mutation of sickle cell allele increases in variation within the population. Having one of these alleles has created a resistance to malaria, therefore populations with a high malaria frequency will also have a high number of people that are sickle cell carriers. The heterozygotes in this population is advantageous because it has a resistance to malaria and they are not affected by the harsh affects of sickle cell anaemia.
4. Individuals that have the sickle cell trait will produce offspring who are likely to inherit either of the genes, therefore passing it onto their children, whereas those who are normal or with sickle cell anaemia will die before reproductive age
5. The allele for the sickle cell trait would eventually become more dominant in the population

THALASSAEMIA

WHAT IS THE DISEASE AND IT'S CAUSES?

Thalassaemia is a genetic disorder that affects the blood. People with thalassaemia do not produce enough healthy haemoglobin (the substance in the blood that enables the red blood cells to carry oxygen around the body). This can make people with thalassaemia anaemic. Thalassaemia is caused by mutations in the DNA of cells that make haemoglobin (chromosome number 11). The mutations associated with thalassaemia are passed from parents to children.

WHAT POPULATION IS AFFECTED BY THIS DISEASE?

It is most common in people of Mediterranean descent as there was once a time marriages between cousins was common among the people inhabiting the countries around the Mediterranean. In Australia it occurs in people of Mediterranean descent, especially Italian and Greek.

WHAT ARE THE MOST COMMON SYMPTOMS OF THIS DISEASE?

- Fatigue
- Weakness
- Pale or yellowish skin
- Facial bone deformities
- Slow growth
- Abdominal swelling
- Dark urine

ARE THERE ANY TREATMENTS FOR THIS DISEASE?

People with thalassaemia require frequent blood transfusions throughout their life and special drugs to remove excess iron that tends to build up in the body. Bone marrow cells produce red and white blood cells, haemoglobin, and platelets. A transplant from a compatible donor may be an effective treatment, in severe cases.

HOW HAS THIS DISEASE PERSISTED IN THE POPULATION?

Due to the small populations there is a greater chance the offspring will inherit two recessive alleles. As it was common to marry between cousins, the allele for thalassaemia has persisted as there a higher chance this allele will maintain within a population as there is no room for gene flow to introduce new advantageous alleles.

WHAT EVOLUTIONARY PHENOMENAS ARE IN PLAY?

Sociocultural barriers - the geographical and cultural isolation of these groups in the Mediterranean meant marriage between cousins became more common. This reduces genetic diversity and also increases the risk of disease within a population

Founder's Effect - deleterious recessive alleles have a higher frequency than in the original population

HUMAN BIO TOPIC 5

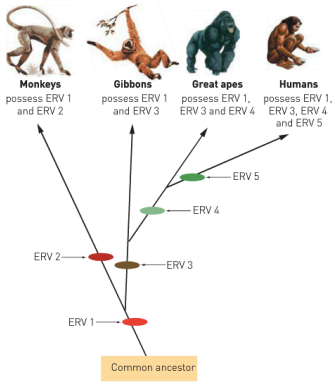
COMPARATIVE BIOCHEMISTRY

COMPARING DNA

DNA is a chemical compound that makes up genes that determine what type of protein a cell can make. All living organisms use the same four bases to code DNA - **guanine, adenine, thymine and cytosine**, which adds weigh to idea that all living things have evolved from a common ancestor.

- Sequence of bases in DNA varies for each species
- New genes gained by mutation, others lost by natural selection or genetic drift
- When speciation occurs, new species have similar DNA, but accumulate more differences overtime
- Species more closely related have less differences in their DNA
- Complete set of DNA in each cell of an organism is called a **genome**
- Humans and chimps share about 98% of their DNA

ENDOGENOUS RETROVIRUSES



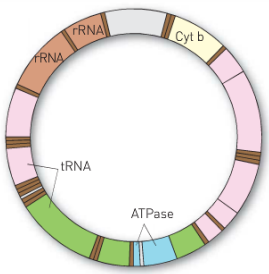
ERVs are sections of non-coding DNA that's a viral sequence that has become part of the organisms genome. It copies its RNA into the cells DNA using reverse transcription and it becomes inserted into hosts chromosomes. Only endogenous if inserted into gamete, so it will be inherited by next generations. Offspring will then have the ERV in exact same location.

- When comparing chromosomes of humans and chimpanzees, same ERVs are located on 16 chromosomes
- Compelling evidence they share a common ancestor

APPLICATIONS OF BIOTECHNOLOGY

1. **PCR** - used to amplify minute amounts of DNA for molecular and genetic analysis. Important when dealing with fossil evidence as only small amounts of DNA may be available.
2. **DNA Sequencing** - to allow comparisons between two species DNA strands
3. **Gel Electrophoresis** - establishes a profile that allows for tracing ancestry and relationships between individuals and groups
4. **Restriction enzymes** - used to cut DNA into smaller fragments to be analysed

MITOCHONDRIAL DNA



Mitochondria are the organelles in the cell were cellular respiration occurs. Small amounts of DNA are found there called **mtDNA**. It is in the form of small, circular molecules. It has 37 genes; 24 code making tRNA molecules and 13 have instructions for making enzymes for cellular respiration.

Inheritance method:

- Egg has many hundred mitochondria, once sperm penetrate egg mitochondria is destroyed
- mtDNA is inverted from mothers
- Similarity of mtDNA estimates closeness of relationship, allow to track ancestry and migration routes
- Has high mutation rate and therefore slowly diverging from mtDNA of original female ancestor

PROTEIN SEQUENCES

Provide evidence for evolution as there are tens of thousands of proteins in living things, all made by 20 different kinds of animo acids. We can see degree of similarity between species by comparing type and sequence of amino acids in similar proteins.

- Animals of same species have identical amino acid sequences and those from different species have different amino acids or order
- Degree of difference enables estimate of amount of evolution that's occurred
- **Ubiquitous proteins** are proteins that appear in all species
- These perform very basic but essential tasks for life, all carry out same function
- Example is **Cytochrome C,** protein that performs essential step in production of cellular energy, changed very little over millions of years
- 37 amino acids are found at same position for every Cytochrome C molecule, suggest descended from ancestor

BIOINFORMATICS

Combines all areas of biological science, using computer science to describe molecular components of living things.

- Enable to compare entire genomics
- Trace evolution of many organisms by measuring changes in DNA

Organism	Estimated size (base pairs)	Chromosome number	Estimated gene number
Human	3.0 billion	46	21 000
Mouse	2.9 billion	40	21 000
Fruit fly	165 million	8	13 000
Roundworm	97 million	12	19 000
Yeast	12 million	31	6 000
Bacteria	4.6 million	1	3 200

COMPARATIVE GENOMICS

Genome sequences of different species are compared to identify similarities and differences. Helps evolution as it identifies preserved genes and unique characteristics of individuals.

- Able to tease apart subtle difference between animal species
- High level of similarity between closely related species
- Reveals diversity of gene composition in different evolutionary lineage

LIMITATIONS TO THE FOSSIL RECORD

The fossil record is incomplete as the **conditions for fossilisation** do not occur regularly. For a fossil to be formed the conditions must require:

1. Rapid burial of the material
2. Presence of hard body parts
3. Absence of decay organisms
4. Long period of stability/undisturbed

Fossilisation is therefore a chance occurrence and there are gaps in fossil record as not every organism is able to be preserved.

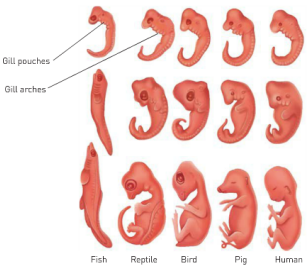
- Only small proportions of organisms that are fossilised are able to be found as some are **inaccessible**
- Some fossils **destroyed** by human activity such as mining
- **Dating** fossils can be problematic such as C-14 can't date older than 60 000 years and K-Ar relies on other suitable materials to be present
- Its unusual to find fossils of the **whole organism**

COMPARATIVE ANATOMY

EMBRYOLOGY

In vertebrates, comparing embryonic stages reveals similarity between different species at different times.

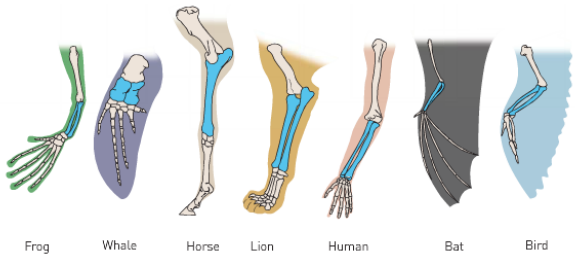
- **Embryonic gill pouches and arches** appear in all species
- Suggests evolutionary series that begun with fish hundred if years ago
- Evolution resulted in **divergence** into amphibians, reptiles, birds etc
- All embryos also have two chambered heart and similar brain structure

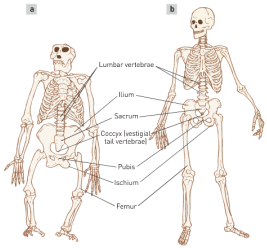


HOMOLOGOUS STRUCTURES

Homologous structures are the same structures that acquire different functions.

- Forelimb bones are **homologous organs** because they posses similar structures but have different functions suited to each species
- Evidence range of vertebrates share common ancestor
- Feet of amphibian, leg of horse, flipper or whale and human hand all have similar structure
- Anthropoids share many anatomical resemblances even though they occupy different habitats

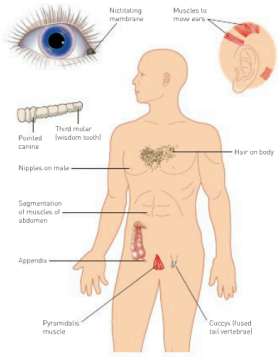




VESTIGIAL STRUCTURES

Are structures of reduced size that appear to have no function when considering their original role.

- Common in vertebrates species and form an intriguing aspect of comparative anatomy
- Can show relationships between species and evidence for evolution
- Example is **coccyx** which was used for a tail and then fused together in humans as it is no longer essential to survival, therefore gradually reduced



GEOGRAPHICAL DISTRIBUTION

Provides evidence for evolution found in the natural geographical distribution of related species.

- Isolated areas have frequently evolved their own distinctive plant and animal populations
- Example is finches on Galapagos Islands that were isolated and developed unique characteristics to those on mainland as different selection pressures were present

FOSSILISATION

A fossil is any preserved trace of an organism that lived long ago; footprints, burrows, impressions, faeces, bones, shell or teeth. They allow scientists to develop picture of the past and provide strong evidence for evolution.

FOSSIL FORMATION

Fossilisation is a chance occurrence as conditions for fossilisation are very specific and micro-organisms decay the fossil before it is found. Conditions in which fossils are formed include:

- **Sand**, **mud** deposits by rivers, volcanic **ash** or other members of the species
- Soil with no oxygen, such as **peat**, preserves soft tissues and bones
- **Alkaline** soils produce best fossils as minerals in bones aren't dissolved
- **Petrification** (turned into rock) when new minerals, like **lime or iron oxide**, replace the organic matter in pores of bone whilst preserving the details of the organism
- Lakes and rivers build up **sediments** when flooding occurs/water flow slows rapidly
- Many **caves** are made of limestone which contains **calcium carbonate** that deposits around dead organisms

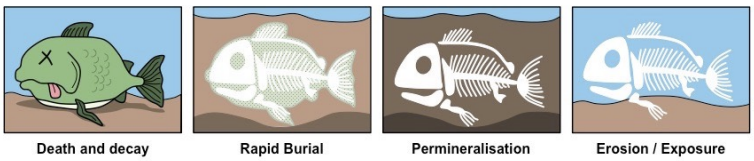
NOTE: WET ACIDIC SOILS DISSOLVES MINERALS IN BONES = NO FOSSILS

DISCOVERY OF FOSSILS

Fossils are sometimes discovered by chance at the surface of ground where they have been uncovered by **erosion**. Surface discoveries are indicators of places that **excavations/digs** may prove fruitful.

- Area is surveyed and marked out in sections
- Small hand tools remove soil gently
- Soil is sieved so no small fragments are lost
- Photographs are taken at every stage
- Fossils are cleaned, pieced together, measured and moulded

Artefacts are objects made by humans such as tools, carvings and paintings that are sometimes dug up with human fossils.



ABSOLUTE DATING

Absolute dating gives the actual age of a specimen (not exact) which allows us to establish the sequence of events that lead to evolution and make discoveries of new species that once existed on Earth.

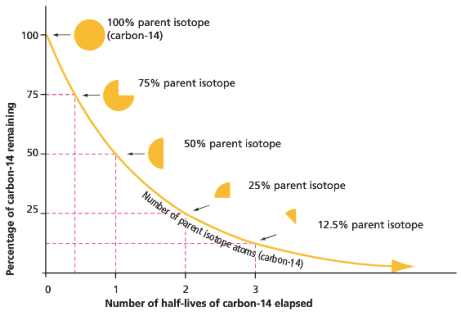
POTASSIUM-ARGON DATING

This method is based on the accumulation of argon in a substance from the decomposition of radioactive potassium.

- Potassium is a mix of three different forms with atomic weight 39, 40 and 41
- Different forms of the same element with different number of neutrons are called **isotopes**
- Isotope **potassium-40** is radioactive and decays to make **calcium-40 and argon-40**
- The actual date is comprised of the time it has been formed from molten/heated minerals
- K-Ar method gives date piece of rock was "reset" by changing its chemical structure via heat/weathering
- By comparing the proportion of K-40 to Ar-40 in a sample of volcanic rock, and knowing the decay rate of K-40, the date that the rock formed can be determined
- Decay of K-40 happens at slow and steady rate
- Easy because argon usually does not leech out of a mineral and is easy to measure in small samples

LIMITATIONS:

1. Suitable rock of the same age must be available
2. Not all rocks are suitable, cannot date bone
3. Only dates rocks older than 100 000-200 000 years old



RADIO-CARBON DATING

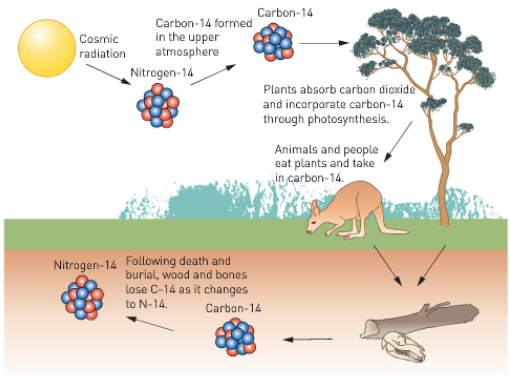
This method is based on the decay or **radioactive isotope Carbon-14** into nitrogen. Carbon-14 is produced by the **cosmic radiation** on nitrogen ions in the upper atmosphere.

- In the atmosphere there is one C-14 to every trillion (10^{12}) stable isotope C-12
- C-14 incorporated into tissues of living things as animals eat plants which get C-14 from photosynthesis
- C-14 decays at a fixed rate with death
- Measuring amount of radiation liberated by a sample, ratio of C-14:C-12 can be estimated and age is calculated
- **Half life** is the time taken for half of any given amount of the isotope to decay, for C-14 its 5730 years until ratio of C-14 to C-12 becomes $0.5:10^{12}$

Accelerator Mass Spectrometry Carbon Dating (AMS) can be used to date samples small as 100mg. It involves breaking sample up into constituent atoms so number of atoms for each isotope can be counted. Useful for dating cave paintings.

LIMITATIONS:

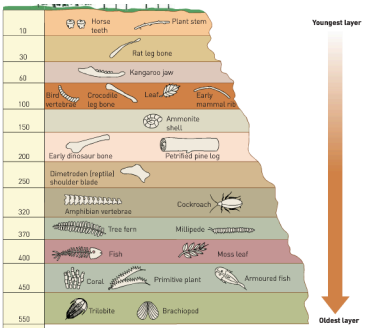
1. Can't be used date back more than 60 000 years
2. Must contain organic compounds (therefore cannot date rock)
3. C-14 in atmosphere has been found to vary
4. At least 3 grams of organic material must be available
5. Not very accurate for fairly recent deposits



RELATIVE DATING

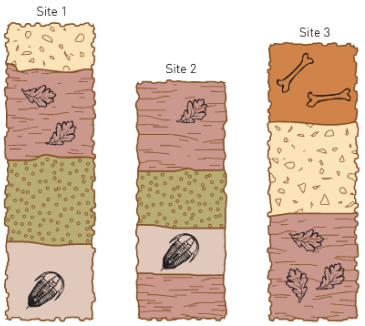
When it is not possible to determine actual age of fossil, we are able to determine if it is older or younger than other samples or the rock/soil in which it is found.

STRATIGRAPHY



Stratigraphy is the study of layers or strata. One of the most fundamental principles of archaeology is the **Law of Superposition:**

- Assumes layers on top are younger than those below
- Sedimentary layers are deposited in a time sequence
- Must be approached with caution as distortion of Earth's crust and burial by animals may change order of strata



The second use of stratigraphy is the **Correlation of Rock Strata** which involves the matching of layers from different areas. This can be done by examining rock itself or the fossils it contains.

- Rocks with the same fossils are the same age
- **Index fossils** are fossils that were on earth for a short period of time and used to correlate strata from different locations
- Index fossils are: widely distributed, only on Earth for short period of time, easily recognisable and abundant
- Analysis of fossilised pollen grains are useful as index fossils and presence of preserved pollen grains in rock/soil sample constructs picture of type of vegetation in area at given time

FLUORINE DATING

Based on the fact that when a bone is left in soil, fluoride ions replace some of the ions in the bone itself.

- Older fossils will contain more fluoride
- All fossil bones in a particular deposit should contain the same amount of fluoride so misplace fossils are identified
- Concentration of fluoride in ground water varies at different areas/times

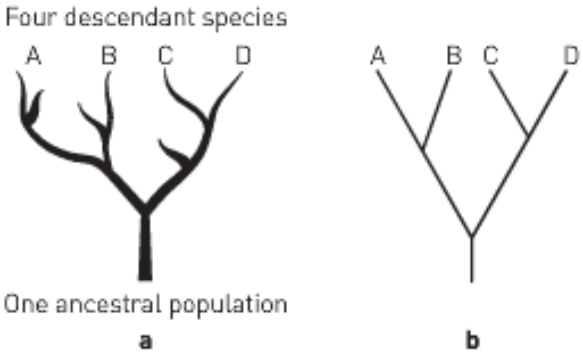
LIMITATIONS OF RELATIVE DATING:

- Simply assuming that an object is older because it was found at a lower depth in the record is only subjective science
- There are many instances of deep holes being dug for rubbish pits or to locate well water that protrude into the record of older strata injecting more modern material as they are filled in over time.
- Landslides and slips can completely change the topography of an entire site burying what was once on top by that which is much older, hence reversing the strata layers

PHYLOGENETIC TREES

Represent the evolutionary relationships between a number of organisms derived from a common ancestor.

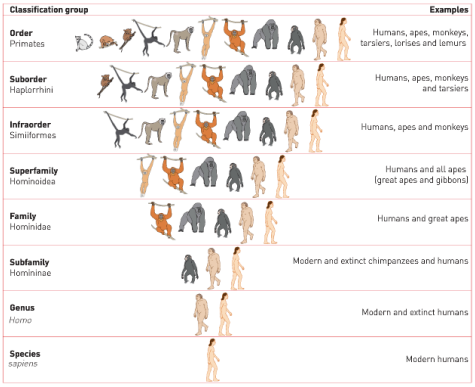
- Ancestral organisms form base of tree
- Those that arise from ancestor are "branches"
- Relationships are shown by distances of organisms



PRIMATES TO HOMININS

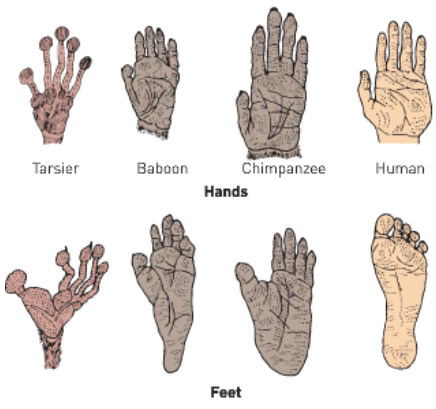
WHAT ARE PRIMATES?

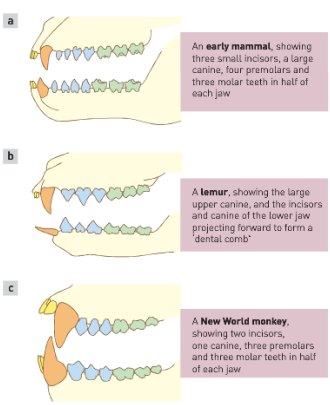
The closest living relative to the human species are classified as part of the Order Primates, including humans themselves. Evidence from the study of protein sequences and DNA has changed the way scientists view relationships between various groups of primates. Humans are classified into the same family as great apes; chimpanzees, bonobos, gorillas and orang-utans, as they share many similar characteristics and DNA.

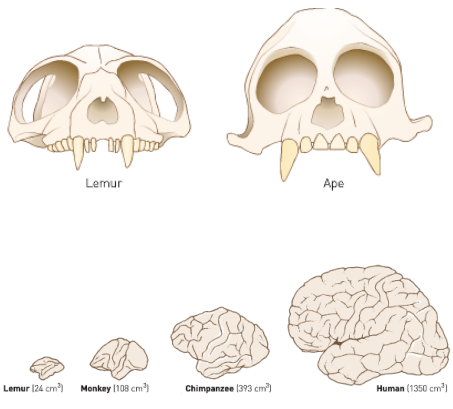


CHARACTERISTICS OF PRIMATES

FEATURE	PRIMATE CHARACTERISTIC
Body	• Not specialised for any particular environment
Limbs	• Generally unspecialised
Hands/Feet	• Pentadactyl - five fingers or toes • Nails instead of claws • Grasping fingers and toes with friction ridges for gripping • First digit opposable
Eyes	• Forward facing for 3D (stereoscopic) vision • Able to distinguish colour
Smell	• Very poor, small olfactory centre
Teeth	• Four incisors in both lower and upper jaw
Brain	• Large and complex • Cerebrum size increases as primates become more highly evolved
Reproduction	• Not restricted to a breeding season • Rhythmical sexual cycle • Usually only one offspring at a time • Long periods of parental car







EVOLUTIONARY TRENDS IN PRIMATES

CHARACTERISTIC		TREND
DIGITS	Mobility	Increasing mobility in digits and ability to move independently, increase prehensility for climbing
	Opposability	First digit opposable to allow for manipulation
	Claws/nails	Nails instead of claws increases grasping technique
	Friction ridges and precision grip	Allows better grip & handle small objects effectively
DENTITION		36 teeth in lemurs, lorises and New World monkeys
		32 teeth in Old World monkeys, apes and humans
		Monkeys and apes have large projecting canines with diastema
		4 cusped molars in monkeys, 5 cusped in apes and humans
SMELL		Sense of smell reduced with gradual reduction in snout
VISION	Eyes	• Increasing efficiency in vision • Gradually forward-facing eyes for stereoscopic vision
	Eye socket	Eyes gradually become enclosed in bony socket to give protection
	Visual area of brain	• Increasing areas of cerebrum devoted to vision • Rods and cones which allow for light and colour
BRAIN	Size	Increase in size relative to body size and complexity
	Convolutions	Gradual increase in number of folds in surface of cerebrum to increase surface area of brain
	Cerebral cortex	Makes up increasingly large proportion of the brain
GESTATION		Increasing length between fertilisation and birth
DEVELOPMENT	Dependance	Increasing length of time offspring are dependant on parents
	Sexual maturity	Later development of sexual maturity

HUMAN ANCESTORS

Humans have the same basic characteristics as apes and therefore are classified into the family Hominidae. Humans, however, have developed characteristics that set them apart from other primates. They belong to the tribe, Hominini, which includes humans and their extinct ancestors.

- From an ancestral ape-like creature, the first hominins evolved; **Australopithecus**
- This evolved early members of the genus **Homo**; **Homo Erectus, Homo Neanderthals** and eventually **Homo sapiens**

ADAPTATIONS FOR BIPEDALISM

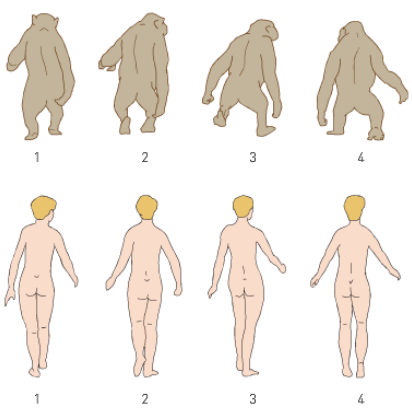
LOCATION	FEATURE	DESCRIPTION	ADVANTAGE
SKULL	Foreman Magnum	Centrally placed at bottom of skull	- Better balance of skull, allows supported by vertebral column - Brings C.O.G over feet
	Prognathism	Flatter face	Allows better balance of skull
	Neck muscles and nuchal area	Muscles and nuchal area are smaller in face	Skull is better balanced so larger muscles aren't required
PELVIS	Size	Shorter in length and wider, bowl shaped	- Supports weight of upper body when standing erect - Supports foetus during pregnancy - Provide larger SA for attachment of buttocks muscles for walking
	Position	Tilted to vertical position	Lowers C.O.G and brings balance over feet
CURVATURE OF SPINE	S-shaped curve	Lumbar curve	- Positions trunk of body over the feet - Carries weight of upper body
		Cervical curve	- Positions head over the neck for smaller spinous processes - Head is forward facing
LEGS AND GLUTES	Glutes	Gluteus muscles modified	Improves hip extension and stride when walking
		Stronger gluteal muscles	Holds body upright
	Femur	Carrying angle	Distributes weight and brings it towards outside of femur, over feet to allow for greater stability
		Larger head of femur	Fits into acetabulum of pelvis to increase stability to carry weight of upper body
		Long legs compare arms	- Longer legs lowers C.O.G which increases stability - Increase stride length when walking - Ability to hold tools whilst walking
	Knee	Strong outer hinge/ condyles	Supports weight due to carrying angle
		Can be straightened	- Allows for striding gait - C.O.G falls infant of knees
FEET	Calcaneous	Larger calcareous bone	- Improves flexion - Takes weight when standing and walking
	Arches	Transversal arch	Shock absorber
		Longitudinal arch	Transfers weight distribution and energy efficiency

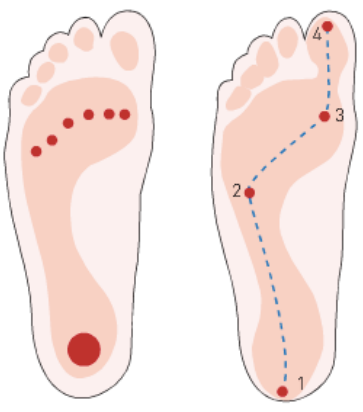
ADVANTAGES AND DISADVANTAGES TO BIPEDALISM

ADVANTAGES	DISADVANTAGES
TALLER: - Able to see over tall grass, spot predators/food, further land is visible - Intimidation of predators/competitors	**TALLER:** - Greater exposure to potential predators
FREE HANDS: - Allows to carry tools, food, infants etc	**FREE HANDS:** - Lifting heavy objects puts strain on back
THERMOREGULATION: - Less surface area exposed to sun's heat - More body exposed to wind for cooling effect	**INJURY:** - If injury to foot or leg, becomes impossible to move - Spine, pelvis and acetabulum, knees and feet still not fully evolved for bipedal walking - Hence problems with pain in these areas
ENERGY EFFICIENCY: - More energy efficient to walk on two legs as less is expended	

STANCE AND LOCOMOTION

FEATURE	DESCRIPTION	ADVANTAGE
MUSCLE TONE	Partial contraction of skeletal muscles	- Allows to keep head erect - Maintains equilibrium of the body
	Nervous system and and sense organs work with spine, hip, knee, ankle and abdominal muscles	- Sustained muscle tone in muscles which support an upright position
STRIDING GAIT	Walking where hip and knee fully straighten	Allows to walk in straight line
	Big toe inline with other toes	Weight is transmitted from heel, to along outside of foot, across ball of foot and then propelled from big toe
SWINGING OF ARMS	Forward swinging of arms compensates for natural rotation of trunk around pelvis	- Less energy is expended - Keeps shoulders at right angle to direction of travel
CARRYING ANGLE	Femurs converge towards knees	- Allows weight distribution to remain close to central axis of body - Stability during walking as body can be rotated about lower leg - Walk in straight line





RELATIVE SIZE OF CEREBRAL CORTEX

FEATURE	DESCRIPTION	ADVANTAGE
LARGE BRAIN SIZE	Human brain size is average of 1350cm whereas apes are between 400cm-500cm	- Increases cranial capacity (volume inside cranium) - Increased thinking capacity and abstract thinking - Higher proportions of cerebral cortex increases mobility
	Convolutions	Give a 50% increase in surface area which gives greater development of frontal lobe
	Frontal lobe	- Higher order of thinking, reasoning, planning and processing - Increase in size and convolutions
	Cerebral cortex	- Have much a larger cerebral cortex area than our direct ancestors and great apes - Site of higher function; vision, memory and reasoning - Allows development of special skills such as tool making
SKULL	Increased rounding of cranium and skull sized	More of skull is used to protect the brain and accommodate for larger frontal lobe
ENDOCASTS	Impression from inside of brain made from rock or other solid material	Reveals trends in number of convolutions and size of frontal lobe

DENTITION AND PROGNATHISM

FEATURE	DESCRIPTION	ADVANTAGE
DENTAL ARCADE	U shaped in primates and more V shaped in humans	
SIZE OF TEETH	Canine teeth do not project beyond other teeth	Changes in diet, no longer needed for intimidation and competition
	Absence in diastema	Allow more space in mouth to articulate speech
	Teeth size and molars reduced	No longer needed due to use in tools and softer foods developments
PROGNATHISM	Flattening of face, development of chin and more prominent nose	Allows frontal lobe to increase in size for higher order of thinking
BROW RIDGE	Sagittal crest disappeared	Humans able to hold neck up without large neck muscles
	Distinct forehead and reduction in brow ridge	Enlargement of cranial portion of brain to accommodate increasing size of frontal lobe

MOBILITY OF DIGITS

FEATURE	DESCRIPTION	ADVANTAGE
DIGITS	Mobility (pentadactyl)	Increasing mobility in digits and ability to move independently, increase prehensility for climbing
	Opposability	First digit opposable to allow for manipulation, longest thumb in relative size to fingers allows for finer manipulation
	Claws/nails	Nails instead of claws increases grasping due to pressure
	Friction ridges	Allows better grip & handle small objects effectively
	Grip	- **Precision grip** allows fun manipulation of small objects using tips of fingers - **Power grip** enables underside of fingers and palm to hold items tight whilst thumb applies pressure in opposite direction

SIMILARITIES AND DIFFERENCES BETWEEN APES AND HUMANS

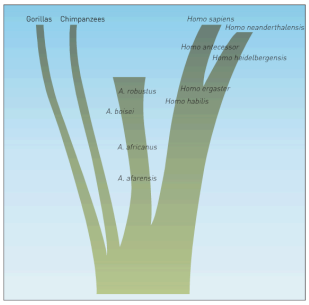
FEATURE	APES	HUMANS
BRAIN	Average 400cm to 500cm	Modern humans average 1350cm
	Cerebrum smaller with fewer convolutions	More convolutions with larger frontal, temporal and occipital lobe
SKULL	More rugged with bony nuchal area and sagittal crests	Smooth and rounded
	Prominent brow ridge	Brow ridge reduced
	Prognathic face	Flatter face
	Large zygomatic arches	Smaller zygotmatic arches
	Large nuchal area for attachment of large neck muscles	Small nuchal area as less muscles needed
DENTITION	Dental arcade is rectangular, U-shaped	Dental arcade is shorter and V-shaped
	Large protruding canines and incisor teeth	Smaller, non-protruding canines and incisor teeth
	Diastema	No diastema
	Chin not developed	Protruding chin in modern humans
SPINE	Robust cervical vertebrae	Robust lumbar vertebrae
	No lumbar curvature	Prominent lumbar curvature
PELVIS	Long and narrow	Wide and short
	Gluteus maximus moves legs side to side	Gluteus maximus swings leg back and forth
LEGS	Arms are longer than legs	Arms and legs are similar length
	Shorter femur with small head	Longer femur with larger head
	No carrying angle present	Carrying angle present
	Wear on inner condyles	Robust outer condyles and bony buttress preventing over-extension of knee
FEET	Smaller calcaneus heel	Calcaneus long and more robust
	Flat-footed, longitudinal arch	Longitudinal and transverse arch
	Opposable big toe	Big toe in line with other toes
HANDS	Fingers long relative to thumb	Shortening of fingers increase relative length of thumb
	Limited mobility of thumb and other digits	Improved precision grip and flexibility
BODY	Body hair long and coarse	Body hair short and fine

HOMININ EVOLUTION

ENVIRONMENT AND EVOLUTION

Geographical, chemical and fossil information is used to paint a picture of what the environment looked like millions of years ago. This helps us understand how evolutionary trends in hominins was influence by **natural selection.**

- After split from chimp lineage, hominins were thought to live in **woodlands** and **forest environments**
- Early hominins had ape-like hands for living in trees
- Upright locomotion may be **due to gaps in forest canopy** due to change in environment
- Believe bipedalism originated in **arboreal ape-like hominins**
- **Temperatures** began to **fall** and **forest areas diminished in size**, leaving patches between trees
- Natural selection favours apes better at bipedal walking around grasslands
    1. Increased vision
    2. Increased size for intimidation
    3. Hands free for carrying objects
    4. Higher reach
    5. Improved cooling



IMPORTANT FOSSIL FINDS

SPECIES	ENVIRONMENT FOUND	IMPORTANT FOSSILS
AUSTRALOPITHECUS AFARENSIS	- Hadar, Ethiopia - Laetoli, Tanzania	- Lucy (first transition between ape and hominin) - 23m trail of Laetoli footprints show foot structure of bipedalism
AUSTRALOPITHECUS AFRICANUS	Taung and Skerfontein, South Africa	Taung Child, first fossil discovered of australopithecine, need for caution as it was Juvenile
PARANTHROPUS ROBUSTUS	South Africa, open Savannah grasslands and woodlands	Kromdraai, South Africa (limestone caves) found large bones
HOMO HABILIS	- Olduvai Gorge, Tanzania - Koobi Fora site, Kenya – "*skull 1470*"	Differences between skull 1470 and earlier skulls make classification difficult, therefore 'early homo'
HOME ERECTUS	- Java, Indonesia - Limestone cave near Beijing - Tools in St. Acheul and Riviera, France - Found throughout Europe, Asia and Africa	- 'Peking man' very similar to Java fossils, however slightly bigger brain on Beijing specimens
HOMO NEANDERTHAL	- Neander valley, Germany - Le Moustier, France	- Skeletons first discovered in Neander Valley - Lived in last of ice ages in Europe
HOMO SAPIENS	- Cro-Magnon, France - Aurignac	- 5 people, animal bones, tools and shell necklace - Aurignacian tools found

STRUCTURAL EVOLUTION

	FEATURE	TREND	
AUSTRALOPITHICUS AFARENSIS **4 to 3 million years ago**	SKULL	Structure	- Point at back and **top of skull** - Zygomatic arch - Foreman magnum under skull
		Cranial capacity	- Cranial capacity of 430cm cubed - Small in relation to jaw size
		Dentation	- V shaped dental arcade - Large canines and incisors - **Diastema present**
		Prognathism	- Prominent brow ridges - Large long jaw - Sloping forehead - **Flared face**
	LIMBS	Hands	Power and precision grip
		Legs	- Fully bipedal - Big toe splayed
	VERTEBRAE, BODY AND PELVIS	Rib cage	- Cone shaped rib cage - Typical hominin curvature in vertebrae - **50kg, males larger than females**
AUSTRALOPITHICUS AFRICANUS **3 to 2 million years ago**	SKULL	Structure	- Zygomatic arch long - Point at back of skull
		Cranial capacity	Cranial capacity 457cm cubed
		Dentation	- V shaped dental arcade - **Reduced canine teeth**
		Prognathism	- **Reduced prognathism** - Large, long jaw - **Smaller brow ridge** - Higher forehead
	LIMBS	Hands	Power and precision grip
		Legs	Fully bipedal
	VERTEBRAE, BODY AND PELVIS	Rib cage	- Cone shaped rib cage - **Males 135cm, females 110cm**
PANRENTHROPUS ROBUSTUS **2 to 1.2 million years ago**	SKULL	Structure	- **Flared zygomatic arches** - **Wide face** - **Sagittal crest present**
		Cranial capacity	Cranial capacity 542cm cubed
		Dentation	- Small canines - Larger pre-molars/molars
		Prognathism	- Reduced prognathism - Flatter forehead - **Huge lower jaw**
	VERTEBRAE, BODY AND PELVIS	Body	- Taller and heavier than gracile forms - 150-170cm talls

	FEATURE	TREND	
HOMO HABILIS **2.4 to 1.4 million years ago**	SKULL	Structure	- Large brain case - **Bulge in Broca's area for speech** - No posterior point or bump - Rounder, elongated head
		Cranial capacity	Cranial capacity 775-600cm cubed
		Dentation	- Parabolic dental arcade - Reduced canines and molars
		Prognathism	- Narrow, long face - Little prognathism
	LIMBS	Hands	- Ape-like arms, long - Slender limb bones - **More robust hands suggest tree climbing**
		Legs	- Femurs longer than Australopithecine
	VERTEBRAE, BODY AND PELVIS	Body	1.3m tall, 40kg with males larger than females
HOMO ERECTUS **1.8 million to 100,000 years ago**	SKULL	Structure	- **Occipital bun** - **Nuchal line at back of skull,** attachment for neck muscles - Thick bones of skull - **Developed Broca's and Wembler's area in brain** - Central ridge, 'midline kneel' - Flat, broad nose - Jaw more compact and shorter
		Cranial capacity	Cranial capacity 1075cm cubed
		Dentation	- Relatively large molars - Parabolic dental arcade, shorter and rounder at front - Modern teeth similar diet to humans
		Prognathism	- Large, flat face - **Low, sloping forehead** - No chin - Slight prognathism - Thick brow ridge
	LIMBS	Legs	- Human-like arch in foot - Thick bones in limbs - Short toes, aligned with one another
	VERTEBRAE, BODY AND PELVIS	Rib cage	Had more modern features
		Pelvis	
	SKULL	Structure	- Long and low cranium - **Occipital bun** - **Depression (suprainiac fossa)** - Zygomatic arches - Robust skull
		Cranial capacity	**Cranial capacity 1486cm cubed**

	FEATURE	TREND	
HOMO NEANDERTHAL **400,000 to 40,000 years ago**		Prognathism	- Sloping/receding forehead - Forward thrust on face - Nasal bone projects forward, wide - **Heavy brow ridge** - Small jaw, no chin
	LIMBS	Hands	Long limbs with heavy and powerful muscles
		Feet	
	VERTEBRAE, BODY AND PELVIS	Rib cage	Barrel shaped chest
		Stature	- **Short stature** - Barrel shaped chest
HOMO SAPIENS **200,000 years ago to present day**	SKULL	Structure	- Shorter skulls from front to back - Large, rounded cranium - Eye sockets large and spaced apart
		Cranial capacity	Cranial capacity 1350cm cubed
		Dentation	- Parabolic dental arcade - Small teeth/molars
		Prognathism	- Higher forehead, frontal lobe developed - Lacks brow ridge - Flat faced, no prognathism - Shortened jaw - Chin present
	LIMBS	Hands	Precision grip
		Legs	Longer femurs that converge towards knee
	VERTEBRAE, BODY AND PELVIS	Pelvis	Broad hips

GRACILE VS ROBUST

FEATURE	GRACILE	ROBUST
Depression in side of skull	Small	Large
Prognathism	More pronounced	Smaller
Sagittal crest	Absent	Present
Forehead	Steep	Flat
Mandible (jaw)	Not robust	Robust
Relative size of canines and incisors	Large	Small
Relative size of molars	Small	Large
Height	120-140cm/Shorter	150-170cm/Larger
Weight	Lighter	Heavier

CULTURAL ADAPTATIONS

SPECIES	DIET AND HUNTING	SHELTERS	LANGUAGE/ART/ CUSTOMS	USE OF FIRE
A. AFARENSIS and AFRICANUS	- **Hunter, forager lifestyle** - Allow to exploit environment - Diet included vegetation, nuts, seeds, insects and eggs - Scavenged meat off dead animals	- Lived in **home bases** - Spent time sleeping in trees	No evidence	No evidence
P. ROBUSTUS	- Large molars meant diet included **large amounts of tough vegetation** - Seeds, nuts and roots included in diet - Small amounts of meat consumed	- Lived in **home bases**	- Lived in **social groups** based off **dominant male** and **several females**	
EARLY HOMO	- Diet consisted of **more meat** in order to obtain complex fats to increase brain size - Food-sharing essential - **Men hunters, females are gatherers** - Ate bone marrow		- Sharing of food marks social structure - Increased sense of **interdependence** - Communication within group and **spoken language developed** - Larynx can't produce proper sounds	
H. ERECTUS	- Smaller molars meant **more cooked foods** - Vegetables and meat - **Skilful hunters** that employed many techniques, such as massive baboon slaughter - **Organised hunts**, drove animas into traps using fire	- Built shelters that enabled them to live in different environments	- Increased communication, cooperation and **complex language** - Increased **social gathering** due to fire	- Helped **keep warm**, keep away **predators** and **stampede animals** - Light extended 'home base' activities such as **tool making** and **butchering carcass** - Cooking meat made **food safer** to eat

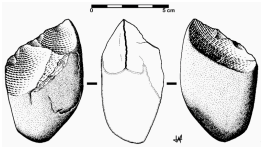
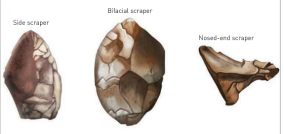
SPECIES	DIET AND HUNTING	SHELTERS	LANGUAGE/ART/ CUSTOMS	USE OF FIRE
H. NEANDERTHAL	- Shared tools with H. Sapiens	- Lived in caves	- **Ceremonial burials** (flowers) - Caring for **disabled members** - Highly developed social system - Made **clothes** - Basic cave paintings	- Used for light and warmth - Stampeding animals
H. SAPIENS	- Animals used for meat, clothing, tools and shelter materials - Hunter/gatherers that relied on **hunting herd animals** - Mastered art of hunting bison, mammoth and reindeer		- Fully **articulate speech** - Cro-Magnon people made **portable art** and **mural art** - Started as line drawing, shading techniques developed - Figurines, statues and decorated tools	

AGRICULTURE

Homo sapiens had great success in colonising a range of geographical areas, which led to the move away from nomadic lifestyles to a more village way of life. This period is called the **Neolithic Revolution or Agricultural revolution**. It was believed to of started 12 000 years ago.

- Fig trees planted as long as 11 300 years ago
- Cereals grown in Syria 9 000 years ago
- Crops, such as wheat and barely, have long since been domesticated and harvested
- **Animal husbandry** followed
- Cattle, goats, sheep and pigs all have origins in farmed animals in area called **Fertile Crescent**
- **Domestication** of these animals from 13 000 to 10 000 years ago
- Cities and civilisations began to grow with agriculture
- No longer had to spend time moving around, could sustain life in one location

TOOL CULTURE

HOMININ	TOOL CULTURE	DESCRIPTION	IMAGE
AUSTRALOPITHECINES	**OLDOWAN TOOLS** - Simple, primitive, no pre-determined design - Made from stones, pebbles	- Basic **pebble tools** - Choppers, scrapers, flakes and chisels - Used to smash open bones - Precision grip must of been employed, round with one edge - Allowed them to **exploit larger areas and therefore migrate**	
HOMO HABILIS		- **Rounded** with **one worked edge** - Cut open prey to obtain flesh quickly - Lived off kills of other animals	
HOMO ERECTUS	**ACHEULEAN TOOLS** - Manufacturing of tools influenced social organisation - Included tools made from bone and stones	- **Hand axes** - **Tear-drop** in shape, flaked all around the edges - Worked on both sides - Helped hunting and building shelters	
HOMO NEANDERTHAL	**MOUSTERIAN INDUSTRIES** - Production of stone flakes that could be trimmed to create various cutting, scraping, piercing and gouging tools	- Stones trimmed into **disc-shaped core** and struck by another piece to produce flakes - Aided in clothes making for cooler climates	
HOMO SAPIENS	**AURIGNACIAN CULTURE** - Used bone and stone to prepare finely crafted tools	- Rectangular 'stone' blade tool with one or two sharp edges - Made by **pressure flaking** - Attached to branches to make axes	
	SOLUTREAN CULTURE - More of a decorative use, served little practical purpose	- **Laurel leaf or willow leaf** shaped flake stone tools - Intricate flaking on every edge - Used as spears or cutting	 **Figure 20.9** A Solutrean 'laurel-leaf' blade
	MAGDALENIAN CULTURE - First tools made by bone, antlers and ivory	- Usually pointed, sometimes with barbs - Included fish hooks, spearheads, harpoons and needles - **Burin**, chisel like cutter was used to make these tools