

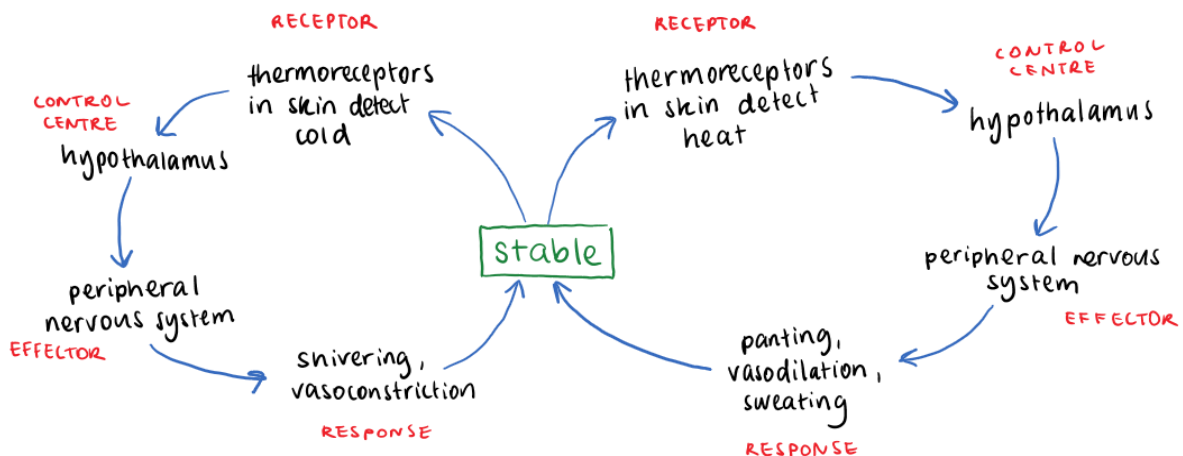
IQ1: How is an organism's internal environment maintained in response to a changing external environment?

1.1 construct and interpret negative feedback loops that show homeostasis, including temperature and glucose

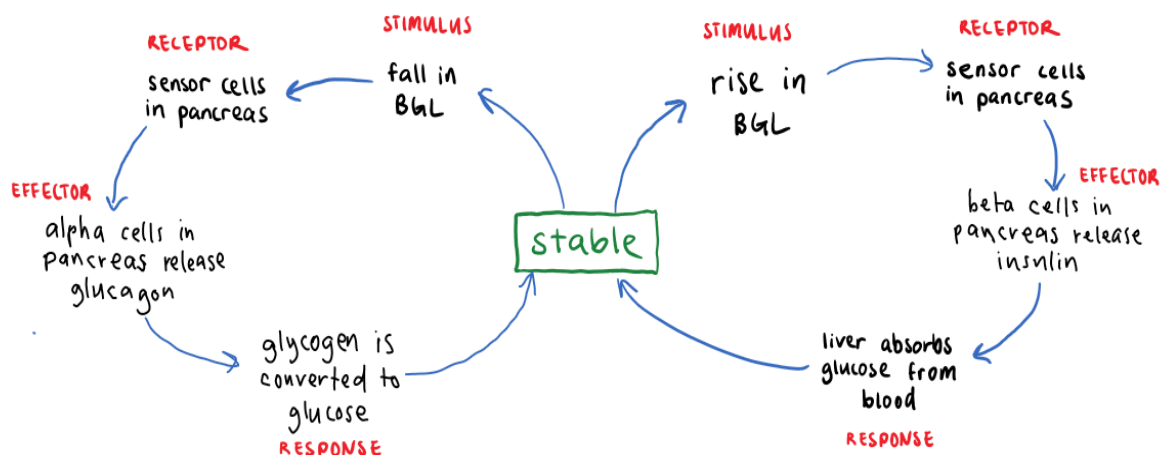
HOMEOSTASIS

- Homeostasis is the maintenance of a stable internal environment despite changes in the external environment.
- Stimulus (change) → receptors (detect change and convert into nerve impulse) → control centre (brain/spinal cord) → messenger (communicates message from CNS to effector) → effector (receives message and causes response) → response (change in body).
- Malfunctions in homeostatic mechanisms can lead to disease.
 - e.g. hyperglycaemia or hypoglycaemia → diabetes
 - e.g. hypothermia or hyperthermia → heat stroke, pneumonia, etc.

TEMPERATURE FEEDBACK LOOP



GLUCOSE FEEDBACK LOOP



1.2 investigate mechanisms used by organisms to maintain their internal environment, including:

– *behavioural, structural and physiological adaptations in endotherms that assist homeostasis*

ANIMAL ADAPTATIONS FOR HOMEOSTASIS

- Heat is exchanged with the environment through conduction, convection, radiation and evaporative cooling.
- Endotherms produce their own body heat through metabolism e.g. mammals and birds
- Ectotherms use the external environment to obtain heat energy e.g. fish, reptiles, invertebrates
- Structural adaptations relate to the size and shape of an organism and its body parts.
 - e.g. kangaroos have network of veins under thin forelimb skin
 - e.g. body proportions suited to climate
- Physiological adaptations relate to how an organism's body works.
 - e.g. temperature regulation – vasoconstriction, vasodilation, sweating
 - e.g. hormonal control of water levels (see: kidneys)
- Behavioural adaptations relate to an organism's behaviours.
 - e.g. snakes seeking sun or shade
 - e.g. kangaroos licking forelimbs to promote evaporative cooling
 - e.g. penguins huddling for warmth

– *mechanisms in plants that allow water balance to be maintained*

PLANT ADAPTATIONS FOR WATER BALANCE

- Xerophytes are plants which are adapted to live in dry conditions – have features that minimise water loss from transpiration.
- Tough cuticles e.g. pigface
 - Thick, waxy surface reduces transpiration
- Fine hairs e.g. paper flower
 - Create humid environment around stomata, reducing water loss
- Water storage e.g. pigface
 - Leaves and stems have large vacuoles to store water
- Extensive root systems e.g. pigface
 - Absorb and store large amounts of water from the soil
- Rolled-up leaves e.g. porcupine grass
 - Leaves curl up when too much water is lost, creating a humid chamber for the stomata
 - Stomata only on one side of the leaf so none are exposed when curled
- No leaves e.g. wattles
 - Flattened stems (phyllodes) instead of leaves allow photosynthesis to occur but lack stomata, reducing transpiration
- Angled branches and leaves e.g. mulga trees
 - Angled down (not horizontal) to direct any water to the roots and minimise amount of sun on leaves
- Transparent leaves e.g. firewheel tree
 - Leaves do not absorb as much heat, slowing water loss

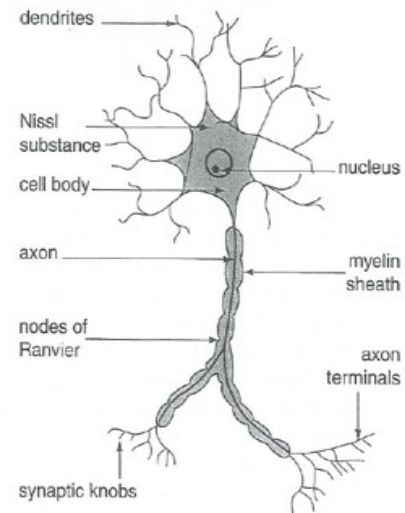
– internal coordination systems that allow homeostasis to be maintained, including hormones and neural pathways

ENDOCRINE SYSTEM

- **Hormones** are signalling molecules that regulate the activity of specific target cells, interacting with specific receptors.
 - Endocrine glands typically release hormones directly into circulatory system e.g. **pituitary gland**

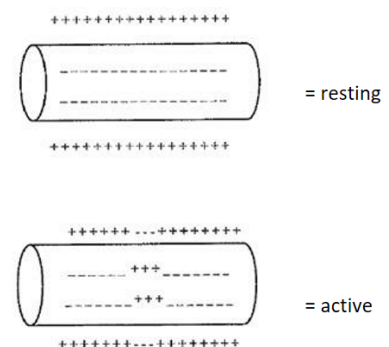
NERVOUS SYSTEM

- Three types of nerve cells: sensory, inter and motor.
 - **Sensory neurones** receive information and send it to the CNS.
 - **Motor neurones** relay signals from the CNS to effector organs (muscles or glands).
 - **Interneurons** connect sensory neurones to motor neurones within the CNS.
- Neurotransmitters e.g. **noradrenaline** are signalling molecules released by nerve cells that communicate electrical signals to chemical signals across a synapse.
 - Presynaptic knobs contain vesicles that release neurotransmitters.
 - Impulses cannot travel across synapses, so neurotransmitters bind to receptors and stimulate dendrites to begin action potential in the next neuron.



ACTION POTENTIALS (NERVE IMPULSES)

- **Action potential** = change in polarity on the inside of the neurone compared to the outside (when nerve is stimulated)
 - i.e. traveling electrical current caused by ions moving through voltage-gated channels.
- Resting potential = charge difference between inside and outside of neurons
 - Sodium ions sit outside, potassium ions sit inside.
- **Steps:**
 - The cell membrane of the neurone is stimulated.
 - The membrane becomes permeable to sodium (Na^+) ions, and they enter.
 - If the threshold (-55) is reached, the membrane becomes more positive on the inside than the outside, creating an action potential.
 - After half a millisecond, the membrane is no longer permeable to Na^+ ions. Potassium (K^+) ions move out, and the resting potential is restored.
 - The resulting depolarisation adjacent to the initial action potential stimulates a new AP as Na^+ ions enter.
 - The AP continues to move along the neurone in this manner.



TYPICAL CHANGES IN MEMBRANE POTENTIAL

Membrane Potential (mV)	Time (msec)	Changes in permeability
-70	0.00	Resting potential
-50	0.75	Threshold
+10	1.00	Depolarisation
+34	1.25	Action potential 'spike'
-60	1.75	Repolarisation
-80	2.00	Hyperpolarisation
-70	3.25	Resting potential

- Threshold = minimum intensity of a stimulus needed to initiate a response.
 - o Intensity is recorded by the rate at which APs are generated → stronger stimuli create more rapid firing and an increase in the number of cells that respond
 - o Speed and strength of APs are constant, it is the FREQUENCY that changes to cause different responses.

IQ2: Do non-infectious diseases cause more deaths than infectious diseases?

2.1 investigate the causes and effects of non-infectious diseases in humans, including genetic diseases, diseases caused by environmental exposure, nutritional diseases, and cancer

INTRO

- A disease is defined as any condition that impairs the body's normal functioning.
- Non-infectious is NOT about contagion – it is about whether or not they can be PASSED ON GENERATIONALLY e.g. germline mutations.
 - Infectious = caused by a pathogen.
 - Contagious = able to be spread from person to person.
 - E.g. tetanus is infectious (soil bacteria) but NOT contagious.

GENETIC DISEASES

- Genetic diseases are caused by changes to the DNA sequence, e.g. by mutation.
 - e.g. haemophilia is a blood disorder caused by a mutation in the blood clotting gene (recessive sex-linked inheritance)
 - Symptoms include frequent/uncontrollable/unexplained bleeding, bruising etc.
 - Treatments include medications to aid blood clotting e.g. plasma or clot factors

ENVIRONMENTAL DISEASES

- Environmental diseases are illnesses caused by factors in the environment e.g. exposure to UV radiation, chemicals, or lifestyle factors such as diet.
 - e.g. lead poisoning is caused by prolonged exposure to lead-contaminated sources, which affects the brain and nervous system (as it is a toxic heavy metal).
 - Symptoms include developmental delays, abdominal pain, neurological changes
 - Treatments include medication to help remove lead from the body
- May be minimised by limiting exposure to these factors and using protective gear e.g. sunscreen.

NUTRITIONAL DISEASES

- Nutritional deficiencies occur when there is insufficient intake of nutrients or the body does not absorb nutrients.
 - e.g. anaemia is caused by insufficient iron intake or inability of the body to absorb enough iron - leads to iron deficiency, and since iron is needed to form red blood cells, the production is slowed.
 - Symptoms include fatigue, fainting, paleness, poor circulation, etc.
 - Treatments include iron supplementation, changes in diet, or iron infusions.

CANCERS

- Cancer involves a group of diseases (skin, breast, lung, etc) in which abnormal cells multiply and spread – i.e. metastasize.
- Cannot be neatly categorised as causes are broad and hard to narrow down.

- o e.g. lung cancer can be caused by environmental exposure such as smoking, however it still occurs in non-smokers.
- o e.g. breast cancer has been linked to a genetic predisposition (BRCA1 and BRCA2), however it may still occur without this.

SUMMARY

Category	Causes	Examples	Effects
Genetic	<ul style="list-style-type: none"> • Gene or chromosomal abnormalities caused by mutation (e.g. by exposure to mutagens) • May be inherited from parents or a result of acquired changes to pre-existing genes 	<ul style="list-style-type: none"> • Huntington's disease • Haemophilia 	<ul style="list-style-type: none"> • Huntington's = degenerative brain disorder: lose ability to walk, talk, think, etc. • Haemophilia = inability to clot blood
Environmental	<ul style="list-style-type: none"> • Exposure to mutagens e.g. lead • Lifestyle factors e.g. poor diet, smoking 	<ul style="list-style-type: none"> • Some cancers e.g. skin, lung • Lead poisoning 	<ul style="list-style-type: none"> • Lung cancer = coughing up blood, collapsed lungs, difficulty breathing • Lead poisoning = organ + brain damage
Nutritional	<ul style="list-style-type: none"> • Insufficient nutrient intake e.g. poor diet • Inability to absorb nutrients e.g. H pylori and iron 	<ul style="list-style-type: none"> • Scurvy (Vitamin C) • Anaemia (iron) • Rickets (Vitamin D) • Osteoporosis (calcium) 	<ul style="list-style-type: none"> • Scurvy = wounds that do not heal, loose/lost teeth • Osteoporosis = brittle bones prone to breakage
Cancer	<ul style="list-style-type: none"> • Genetics e.g. family history • Lifestyle factors e.g. smoking • Environmental factors e.g. sun exposure 	<ul style="list-style-type: none"> • Lung cancer • Skin cancer • Breast cancer 	<ul style="list-style-type: none"> • All = pain, headaches, abnormal bleeding, tumours, etc.

2.2 investigate data to show the incidence, prevalence and mortality rates of non-infectious diseases, including nutritional diseases and environmental diseases

ENVIRONMENTAL DISEASE – MELANOMA

Feature	Description of environmental disease
Name	Melanoma (most lethal type of skin cancer)
Cause	<ul style="list-style-type: none"> • Uncontrolled skin cell division due to changes in DNA that control cell division (e.g. in melanocytes which produce melanin) <ul style="list-style-type: none"> o Affected by exposure to UV radiation e.g. sunlight, age, sunburns o Genetic component → individuals with fair skin more prone
Symptoms	<ul style="list-style-type: none"> • Tumours/growths • Changes in existing moles e.g. getting darker or larger • Swollen lymph nodes • Changes in skin pigmentation
Host response	<ul style="list-style-type: none"> • Primarily inflammatory response/second line of defense • Cytotoxic T cells also move to tumours to destroy them (cells send SOS signals)
Treatment	<ul style="list-style-type: none"> • Surgery (removal of melanoma and surrounding cells) • Chemotherapy • Radiotherapy
Incidence	<ul style="list-style-type: none"> • Approx. 35 per 100,000 in 2017 • Most common cancer in young Australians • Rates are remaining constant
Prevalence	<ul style="list-style-type: none"> • Approx. 49 per 100,000 in 2016 • Prevalence is higher in males than females • Overall VERY high in Aus due to outdoor lifestyles and hole in ozone (more UV)
Mortality rate	<ul style="list-style-type: none"> • Approx. 49 per 100,000 in 2016 • Approx. 1900 deaths in 2018

NUTRITIONAL DISEASE – KWASHIORKOR

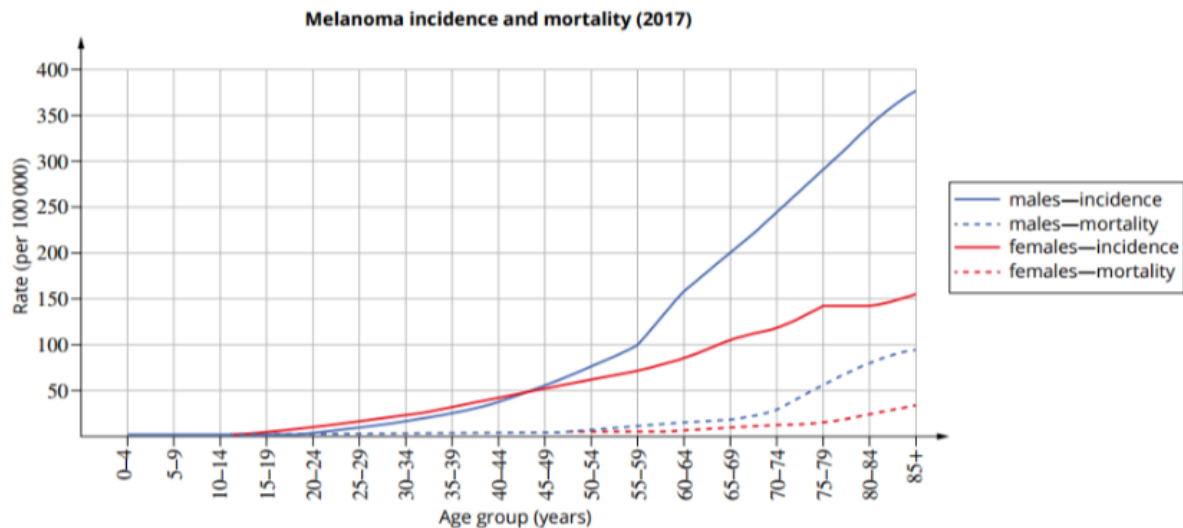
Feature	Description of nutritional disease
Name of disease	Kwashiorkor (the word come from Ghana and means the sickness the older child gets when the next baby is born).
Cause	Severe deficiency of protein in the diet (when next baby is born the older child does not get any breast milk and may have no protein in their diet and so get the disease kwashiorkor).
Symptoms	Oedema – fluid retention with bloating of the ankles and feet, the liver enlarges with fatty infiltrates, fluid is lost from the bloodstream and the abdomen swells. Muscles become wasted and the person feels constantly tired and listless. The hair thins and can change colour and lose its curliness, teeth fall out, the skin develops dermatitis becoming flaky with rashes and there is loss of skin pigmentation. The immune system is affected leading to more frequent and severe infections and there are behavioural changes with increased irritability. Long term effects can show in lower physical and mental development.
Host response	Reduced protein intake reduces plasma proteins, e.g. albumin, fibrinogen and gamma globulins which impairs the body's ability to maintain osmotic pressure causing swellings and lack of antibody production reduces the functioning of the immune system.
Treatment	Treatment involves adding protein to the diet in small rationed doses with essential elements, minerals and vitamins. If treatment is delayed the person can go into coma, shock and major organ failure will lead to death.
Incidence	In Australia kwashiorkor has low incidence and very low mortality rate.
Prevalence	Globally kwashiorkor is most prevalent in overpopulated regions, e.g. Africa, Central and South America and South Asia and is typically seen in young children, e.g. 1 to 3 years old. In Australia malnutrition is a serious problem and has been called the 'silent epidemic'. Prevalence occurs in aged care facilities and isolated rural areas.
Mortality rate	In areas where kwashiorkor is prevalent there is a relatively high case mortality rate.

IQ3: Why are epidemiological studies used?

3.1 analyse patterns of non-infectious diseases in populations, including their incidence and prevalence, such as diseases caused by environmental exposure

PATTERNS OF DISEASE

- Epidemiology is the study of the causes, effects and patterns of disease in populations.
 - Aids in the management, evaluation and planning of services for prevention, control and treatment of diseases.
 - Can be collected from hospitals, insurance claims, GPs, health surveys, etc.
- Indicators include incidence, prevalence, morbidity, mortality and life expectancy.
 - Incidence = number of NEW cases of a disease in a population
 - Prevalence = the PROPORTION of affected individuals in a population (used to measure morbidity).
 - Mortality = number of deaths from the disease
- **EXAMPLE: 50-year study in the UK and US (beginning in the 1950s) to determine the link between smoking and lung cancer → results showed the test group had much higher incidence than the control group, confirming the link.**



3.2 investigate the treatment, management, and possible future directions for further research of a named non-infectious disease caused by environmental exposure

TREATMENT, MANAGEMENT AND FUTURE DIRECTIONS

- Treatment and cure of a disease will depend on the type of disease.
 - e.g. treatment of scurvy involves adding appropriate amounts of Vitamin C to the diet.
 - Other diseases, e.g. cancer, require more complex treatments.
- Screening, early diagnosis and management of symptoms is the best way to deal with diseases that have limited treatment options.
 - e.g. CF currently has no successful treatment → management strategies concentrate on reducing symptoms
- Research into many different types of diseases, how they manifest in the body, and the way systems in the body work contribute to more successful treatments.
 - e.g. research is currently being undertaken into the use of gene therapy to treat CF.

CASE STUDY: MELANOMA

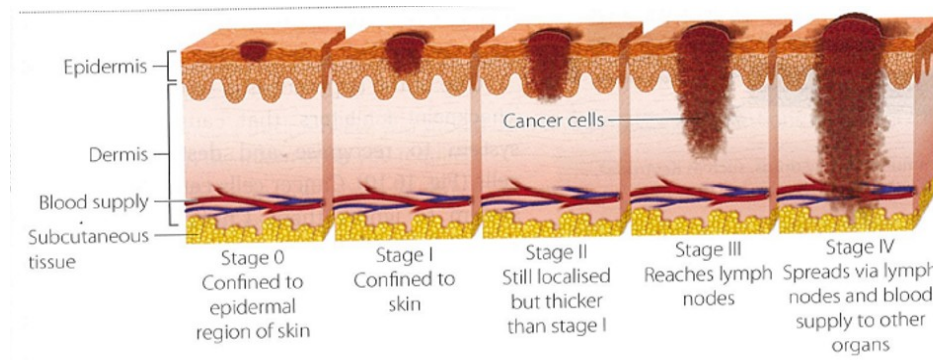


FIGURE 16.9 The stage at which melanoma is diagnosed determines the best treatment option.

Treatment	Description
Surgery	<ul style="list-style-type: none"> • Involves removing the tumour and surrounding skin to ensure that no cancerous cells are left behind. • Most common form of treatment for early-stage melanoma.

Radiation	<ul style="list-style-type: none"> • X-rays damage the DNA of cancer cells and kill them, whilst it allows normal cells to repair their DNA more easily. • Can be administered externally or by placing a radioactive source in the body near the cancer cells. • <i>*Care must be taken in directing radiation to specific cells needing destruction.*</i>
Chemotherapy	<ul style="list-style-type: none"> • Chemotherapy drugs slow the growth of cancer cells. • However, this has not been particularly effective in treating melanoma – used more for other cancers.
Targeted therapy	<ul style="list-style-type: none"> • Involves the use of drugs that affect molecules that control the growth of tumour cells, stopping it spreading. • Melanoma cells have mutations that cause uncontrolled cell division – targeted therapy interrupts the pathways that cause this.
Immunotherapy	<ul style="list-style-type: none"> • Cause the body's own immune system to fight the melanoma – can be done with checkpoint inhibitors or vaccinations. • Checkpoint inhibitors = the use of drugs that cause the immune system to recognise and destroy melanoma cells (since cancer cells cause the immune system to ignore them). • Vaccinations = an antigen is produced using the melanoma cells, and when used, it allows the immune system to more easily identify + destroy the cancer.

- Future research is required for melanoma, specifically in the development of targeted therapies and immunotherapy treatments.
- Research for targeted therapies:
 - Since this treatment is based on different mutations that cause uncontrolled cell division, further research is required to identify all other mutations and develop more drugs.
- Research for immunotherapy:
 - Immunotherapy drugs are not successful on all patients, so more research is required to develop a greater variety for maximum benefit.
 - Research will involve refining the process of vaccinations to make them consistently effective, and investigating the relationship between melanoma and other cancers.

3.3 evaluate the method used in an epidemiological study

EPIDEMIOLOGY QUESTIONS

- Why conduct one?
 - To discover patterns that establish links between cause and effect
 - To develop programs to prevent and control disease.
- What is looked at?
 - Who gets sick and why?
 - What is the most effective treatment?
- What is it used for?
 - To develop programs e.g. vaccination programs
 - To identify people at risk
 - To establish where money must be allocated

TYPES OF EPIDEMIOLOGICAL STUDIES

- Descriptive studies provide the who, what, where and when → generate hypotheses about causes of disease.
- Analytical studies test hypotheses and provide the why or how → determine cause of disease.
- Intervention studies are used to test the effectiveness of a disease treatment or public health campaign.

STAGES OF COLLECTING DATA

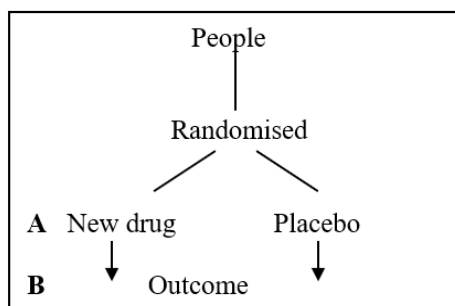
- Diagnostic phase → confirms that disease is present
- Descriptive phase → describes population at risk and distribution of disease
- Investigation phase → implementation of field studies
- Experimental phase → experiments performed in controlled conditions to test hypotheses
- Analytical phase → results are analysed
- Decision making phase → knowledge of epidemiology of disease is used to explore options available for management

TREATMENT VS. MANAGEMENT

- **Treatment** = lessening symptoms/effects of disease
 - Can include cure e.g. antibiotics for chest infection
 - If incurable, helps control e.g. HIV and PrEP
- **Management** = broader picture dealing with prevention, control and communication
 - Can also overlap with treatment, including managing individual symptoms
 - e.g. bowel cancer → treatment involves surgery/chemotherapy, management involves public education and bowel cancer test kits

HOW STUDIES ARE DONE

1. Randomised Trials



e.g. new drug group = hopefully increased life expectancy
control group = no drug (placebo)

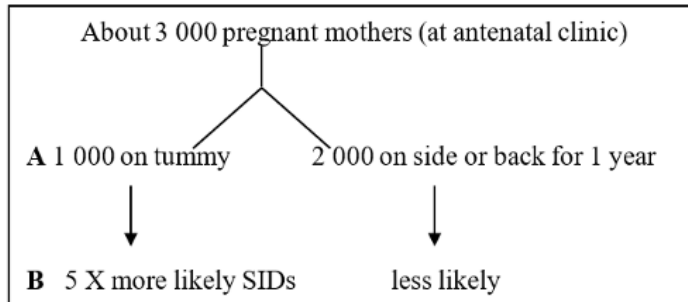
The groups chosen should equal ages and sex and numbers must be high!

All people tested are kept as controlled as possible (e.g. all doing tai chi, same diet, mammograms etc)

2. Cohort Studies

- Used when not practical to do a trial OR if the expected outcome is harmful (e.g. you can't force people to smoke-unethical)

e.g. SIDS observational study (Sudden Infant Death Syndrome) where babies were monitored during and after birth and mothers were asked to monitor babies' sleeping habits weekly.



Results: still don't know the cause but education has decreased the incidence of SIDS **BUT** we cannot be 100% certain as there could be another factor.

****following over time****

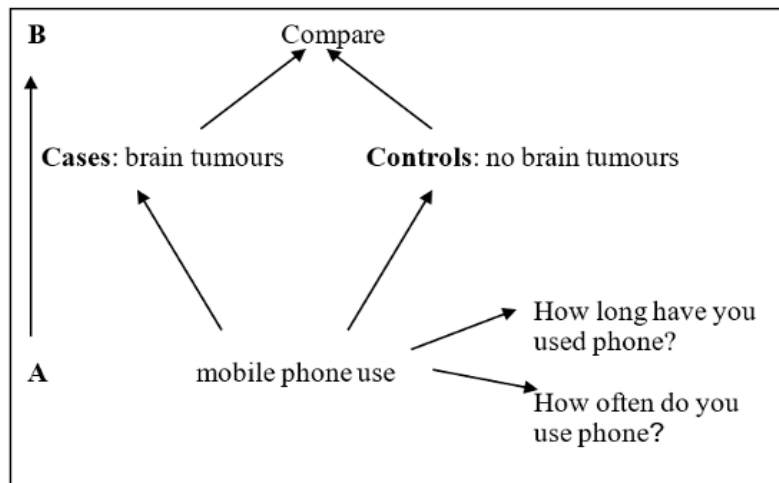
3. Case-Control Study (i.e. flashback study - looking at the history of people reporting issues)

- Start with a group who have the outcome and a group who don't.

e.g. brain tumours and link to mobile use

NB. Brain tumours are rare, and this means very large groups are required.

For mobile phone use the other types of trials can't be used (randomised or cohort because almost impossible to keep all other factors constant).



ERRORS IN EPIDEMIOLOGICAL STUDIES

- Random errors are unpredictable variations in data.
 - Make the study less precise but do not shift the results in a particular direction.
 - Can be corrected using statistics.
 - Usually occur because of differences in subjects being studied → ensuring homogenous groups will reduce Res
- Systematic errors (bias) involve any process during a study that causes a consistent deviation in measurement from what the true value should be.
 - Result in an incorrect estimate of the effect of exposure to a particular factor and the cause of a disease.
 - Selection bias is bias in selecting subjects to include in the study → must be representative of population

- Sampling bias = the way subjects are chosen/where from is not representative of population
- Volunteer bias = those who volunteer already have an interest in the condition and may be at higher risk
- Healthy worker bias = employed people are usually healthier than unemployed
- Prevalence/incidence bias = only current cases are included in the study.
- o **Information bias** is errors in taking measurements or recording information → affects each study group differently
 - Misclassification bias = some subjects already suffer but are undiagnosed
 - Recall bias = those affected by the condition will have greater recall than those who are not
 - Loss to follow up bias = not all subjects are followed up to the same degree
 - Interviewer bias = questions indirectly lead study participants to the answer
 - Measurement bias = measures are consistently inaccurate
- o A **confounding factor** is a type of systematic error that occurs when an unrecognised factor may be affecting the results of the study e.g. studying lung cancer in asbestos workers but ignoring cigarette use
- Errors can be reduced by:
 - o Using statistics
 - o Using multiple observers
 - o Taking repeated measurements
 - o Establishing standards/objective definitions beforehand
 - o Large sample sizes
 - o Using controls
 - o Long periods of study

3.4 evaluate, using examples, the benefits of engaging in an epidemiological study

BENEFITS OF USING EPIDEMIOLOGY

- Identify causes of disease e.g. smoking and lung cancer
- Allows public health authorities to manage, evaluate and plan to control diseases → governments then allocate resources, develop policies and health promotion strategies to improve health e.g. National Tobacco Strategy
- Identify treatments and/or cures for diseases e.g. HIV and PrEP
- Predict possible future outbreaks

IQ4: How can non-infectious diseases be prevented?

4.1 evaluate the effectiveness of current disease prevention methods and develop strategies for the prevention of a non-infectious disease, including educational programs, campaigns and genetic engineering

PREVENTION

- Prevention encompasses many activities:
 - o Reducing the likelihood that a disease or disorder will affect an individual
 - o Interrupting or slowing the progress of an already existing disorder
 - o Reducing disability.

- e.g. mammograms do not prevent breast cancer, BUT it is a *preventative action* because early detection can stop/slow the disease.
- e.g. wearing sunscreen does not prevent melanoma, but reducing UV exposure reduces the likelihood that the disease will affect an individual (1981 Slip Slop Slap campaign).
- e.g. HIV and PrEP -> effective in stopping symptoms, multiplication of the virus and transmission
- e.g. National Tobacco Strategy -> highly successful

EVALUATION OF ANTI-SMOKING CAMPAIGNS

What disease(s) does this help prevent?	Smoking-related cancers e.g. lung cancer and respiratory diseases e.g. COPD
What is currently being done?	<ul style="list-style-type: none"> • PDHPE syllabus • Legislation banning advertising for smoking • Legislation banning smoking in public areas • Legislation requiring warnings on cigarette packets • Heavy taxation on cigarettes to discourage purchase • Funds provided by the government for research • Intersectoral collaboration to encourage quitting e.g. Quitline
Evaluate the effectiveness of current disease prevention methods in this area.	<ul style="list-style-type: none"> • + Very effective -> seen a large reduction in the incidence/mortality of lung cancer in Australia • + Less people are buying cigarettes esp. younger people and boys • + Increased awareness means more people are getting scanned/tested, which means more people are identifying the disease early, increasing the chances of survival and cure • - However heavy taxation is not effective for addicts and is negatively affecting low SES people
Suggest improvements that could be made	<ul style="list-style-type: none"> • Complete ban/outlaw of tobacco • Putting tax into supporting programs for addicts (like low SES) • Raise legal age for purchasing cigarettes to 21 • More legislation

GENETIC ENGINEERING

- Genetic engineering is the deliberate modification of an organism's DNA.
 - o e.g. golden rice = highly effective for Vitamin A deficiencies
 - o e.g. CRISPR Cas9 = gene editing technology for embryos
 - o e.g. pre-implantation screening (IVF) = can prevent offspring inheriting CF
 - o e.g. GE insulin using recombinant DNA technology = highly effective for diabetics

EVALUATION OF GE INSULIN

- Process:

- Human DNA is extracted from pancreas cells, and the insulin-producing gene is isolated using specific restriction enzymes.
- Plasmid DNA (vector) is extracted from a bacterium and cut with the same restriction enzyme.
- The human insulin-producing gene is inserted into the plasmid vector to form recombinant DNA.
- The recombinant DNA plasmid is inserted into a bacterium cell to form a recombinant/transgenic bacterium.
- The recombinant bacterium multiplies in a fermentation tank, producing insulin.
- The insulin is extracted, purified and bottled for use.
- Benefits of GE insulin:
 - The insulin produced cannot trigger an immune response, because the hormone is identical to naturally-produced insulin in the body
 - Brings about a much more rapid response than cow insulin because of the exact fit of insulin receptors in human liver cells
 - Reduces ethical issues e.g. objections from vegetarians

GENE THERAPY

- Gene therapy is an experimental technique that manipulates genes in order to treat and prevent disease at its source, i.e. at gene level.
 - Can be used to modify genes or replace faulty genes with a healthy version.
 - However it only targets body cells, not gametes – so it is not passed onto offspring.
 - Can help to treat incurable conditions e.g. genetic diseases like haemophilia and CF.
 - CF is caused by a faulty CFTR gene mutation that causes a build-up of mucus in the lungs (often lethal) → gene therapy aims to replace some of the faulty genes in the cells with healthy ones.
 - The missing genes are placed inside a modified virus that cannot reproduce.
 - The virus is inhaled by the patient to directly target/insert the gene into lung cells (the virus produces proteins that prevent/control CF symptoms).
 - The virus is also encased in a fat bubble, which stops the body's immune response from destroying the virus. It is usually delivered by a nebuliser.

IQ5: How can technologies be used to help people who experience disorders?

5.1 explain a range of causes for disorders by investigating the structures and functions of relevant organs, then:

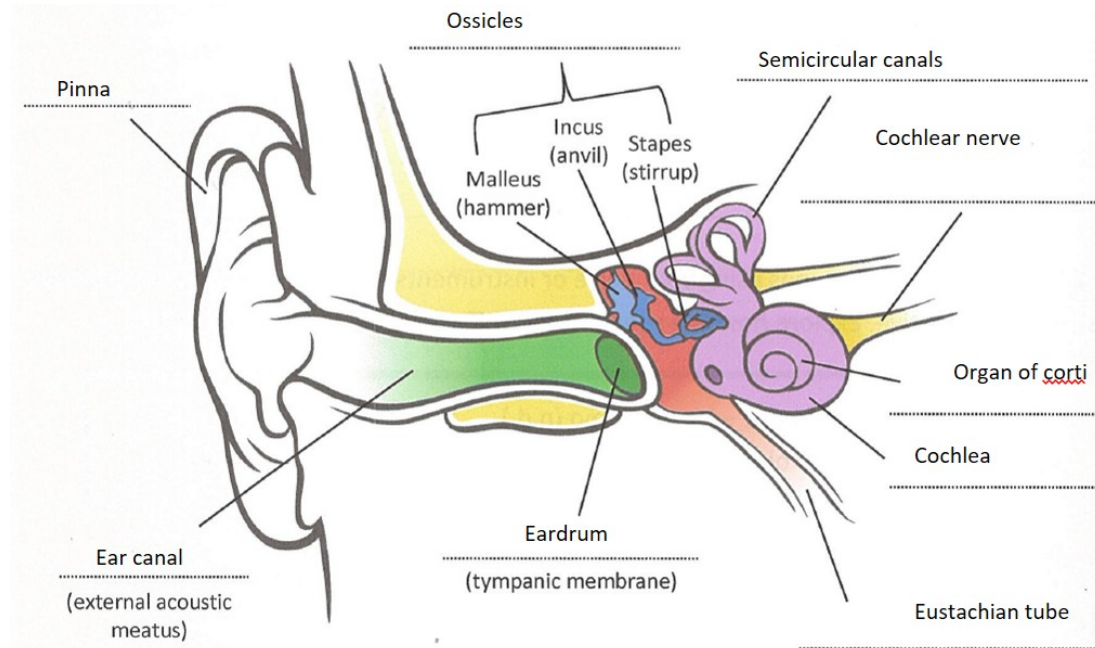
5.2 investigate technologies that are used to assist with the effects of these disorders

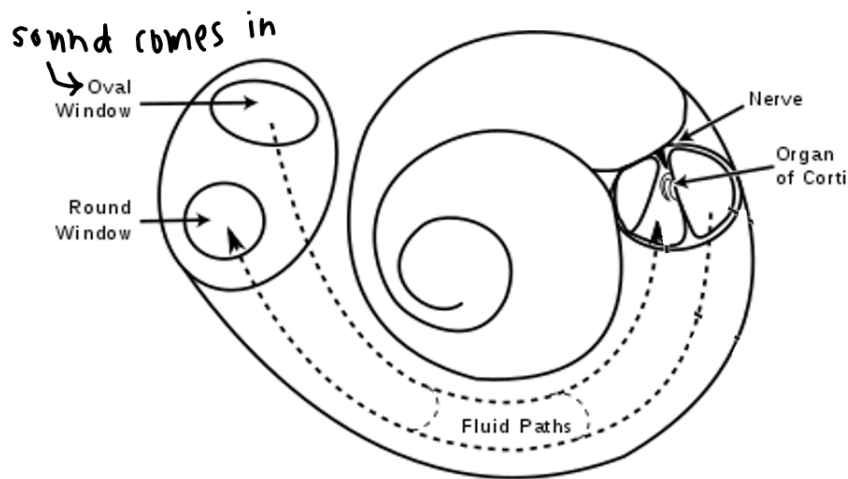
DISORDERS

- Disease and disorder are often used interchangeably, but they are DIFFERENT.
- Disease = a condition in which the body's normal functioning is impaired.
- Disorder = a disturbance that affects the mind or body.
 - o Due to intrinsic abnormalities (differences in an individual's genome) e.g. ADHD
 - o Disorders can be caused by trauma, environmental factors, genetics or naturally through aging.

– hearing loss: hearing aids, bone conduction implants, cochlear implants

STRUCTURE OF EAR





FUNCTION OF EAR

Structure	Function
Pinna	<ul style="list-style-type: none"> External fleshy part of the ear that funnels sound waves into the ear canal
Ear canal	<ul style="list-style-type: none"> Earwax is produced here to help reduce bacterial infections Funnels sound waves to the eardrum
Ear drum/tympanic membrane	<ul style="list-style-type: none"> Membrane at the end of the ear canal that vibrates at the same frequency as soundwaves Transfers sound waves from the external ear into the middle ear
Ossicles (hammer, anvil, stirrup)	<ul style="list-style-type: none"> Three small bones in the middle ear that AMPLIFY the vibrations from soundwaves The stirrup presses against the oval window, passing the vibrations into the inner ear
Eustachian tube	<ul style="list-style-type: none"> Connects the ear to the nose and throat Helps to equalise pressure in the ear + drain fluid
Organ of Corti	<ul style="list-style-type: none"> Situated in the middle canal of the cochlea Contains tiny mechanoreceptors (hair cells) that detect sound vibrations and convert these into electrical impulses
Cochlear nerve	<ul style="list-style-type: none"> Sends electrical impulses to the brain
Round window	<ul style="list-style-type: none"> Vibrates with opposite phase to the oval window as the cochlear fluid is displaced

outer

middle

inner

PROCESS OF HEARING

- Pinna funnels sound waves into the ear canal.
- Sound waves travel down ear canal to eardrum and cause it to vibrate at the same frequency.
- The mechanical vibrations of the eardrum cause the ossicles to vibrate and amplify the sound.
- The vibrations are then transferred to the fluids within the top canal of the cochlea.
- The vibrations passing through the top canal push the fluid in the middle canal (organ of Corti).
- The hair cells (mechanoreceptors) in the organ of Corti detect these vibrations, generating electrical nerve impulses.
 - High frequencies are detected by cells nearest the oval window.
 - Low frequencies are detected by cells furthest from the oval window.

- The impulses travel via the auditory nerve to the brain, where the sound is interpreted.

TYPES OF HEARING LOSS

- **Conductive hearing loss** = caused by blockage or damage in the outer and/or middle ear.
 - o Leads to a loss of loudness → causes include:
 - Blockages of the ear canal e.g. wax or foreign objects
 - Perforated eardrum e.g. from trauma (pencil)
 - Otosclerosis (bone grows around stirrup bones)
- **Sensorineural hearing loss** = caused by damage/malfunction of the cochlea or auditory nerve
 - o Leads to a loss of loudness and/or lack of clarity → causes include:
 - Ageing process
 - Excessive noise exposure
 - Diseases e.g. rubella
 - Congenital abnormalities
- **Mixed hearing loss** = problems with both the conductive AND nerve pathway.

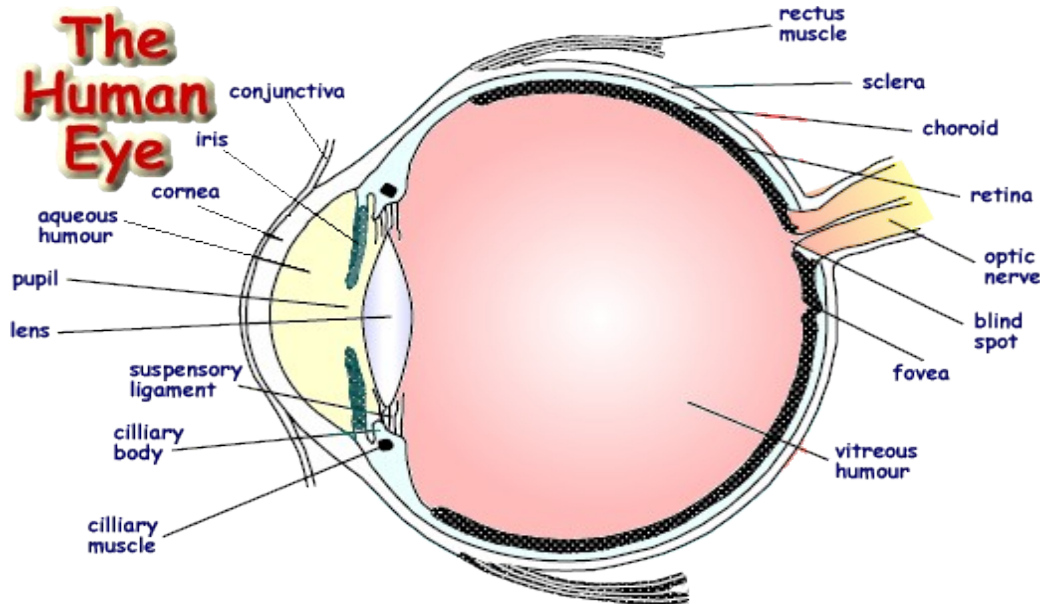
TECHNOLOGIES TO ASSIST HEARING LOSS

- **Hearing aids** = electronic devices that can amplify sounds entering the external ear.
 - o Can help with conductive or mild sensorineural hearing loss e.g. in the elderly
 - o Process:
 - Microphone → detects sound and converts it to electrical signal
 - Amplifier → magnifies/increases strength of sound
 - Receiver → converts electrical signal back into sound
 - Speaker → transmits the sound into the ear
 - o Benefits:
 - Can be programmed to dull background noises, increasing quality of sound
 - Do not require surgery
 - Relatively cheap
 - o Limitations:
 - All parts of the ear must be functioning (to an extent) for it to work
- **Bone conduction implants** = external processors that send sound as vibrations through bone directly to the inner ear
 - o Can help with more severe hearing loss, bypassing both the outer and middle ear
 - o Process:
 - Processor → detects and amplifies soundwaves, then converts them to vibrations
 - Implant → transmits vibrations directly through bone to the cochlea
 - o Limitations:
 - Requires surgery
- **Cochlear implants** = 'bionic ear' sends sound as electrical impulses directly to auditory nerve
 - o Can help to treat severe to profoundly deaf people with missing/damaged hair cells
 - o Process:
 - Processor → detects sound and converts into digital code
 - Implant → converts digital code into electrical impulses and transmits to electrodes in the cochlea
 - Electrodes → stimulate auditory nerve, signals then recognised by the brain
 - o Benefits:

- Can facilitate hearing in profoundly deaf people, improving QOL
- o Limitations:
 - Do not work if auditory nerve is damaged
 - Very expensive
 - Requires surgery
 - Patients with implants have to be trained to interpret sound
 - Do not fully reproduce normal sound (bad with timbre and pitch)

– visual disorders: spectacles, laser surgery

STRUCTURE OF EYE

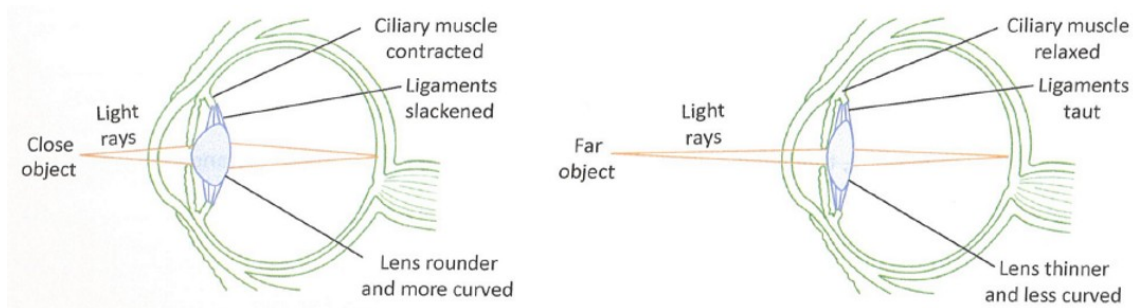


FUNCTION OF EYE

Structure	Function
Sclera	<ul style="list-style-type: none">• Tough white coating that covers and protects the eyeball
Iris	<ul style="list-style-type: none">• Coloured part of the eye that contracts or relaxes to change the size of the pupil in different light levels
Pupil	<ul style="list-style-type: none">• Opening in the middle of the iris that allows light to enter the eye
Cornea	<ul style="list-style-type: none">• The transparent, front surface of the eye covering the iris and pupil• Light refracts most through this layer as it enters the eye
Conjunctiva	<ul style="list-style-type: none">• Thin, translucent membrane covering the sclera
Lens	<ul style="list-style-type: none">• Transparent, bi-convex disc that focuses light onto retina• Can change shape for near and far objects, controlled by the ciliary muscles
Retina	<ul style="list-style-type: none">• Sensory membrane lining the back of the inner eye• Contains photoreceptors and nerves that allow light impulses to be changed into electric signals
Optic nerve	<ul style="list-style-type: none">• Carries electric signals from the retina to the brain
Choroid	<ul style="list-style-type: none">• Supplies blood to the eye
Photoreceptors (rods and cones)	<ul style="list-style-type: none">• Rods receive grey and black EM waves• Cones receive colour (RGB) EM waves and provide clarity of vision
Fovea	<ul style="list-style-type: none">• Central area of retina with greatest concentration of cones, thus the best clarity

PROCESS OF SIGHT

- If light is travelling from a distant object, the light rays tend to be parallel.
 - Less refraction needed = lens at rest + long focal length.
- Light rays travelling from a close source tend to diverge.
 - More refraction needed = lens at maximum accommodation + short focal length.
- **Accommodation** is the focusing of objects at different distances, caused by changing the convexity of the lens.

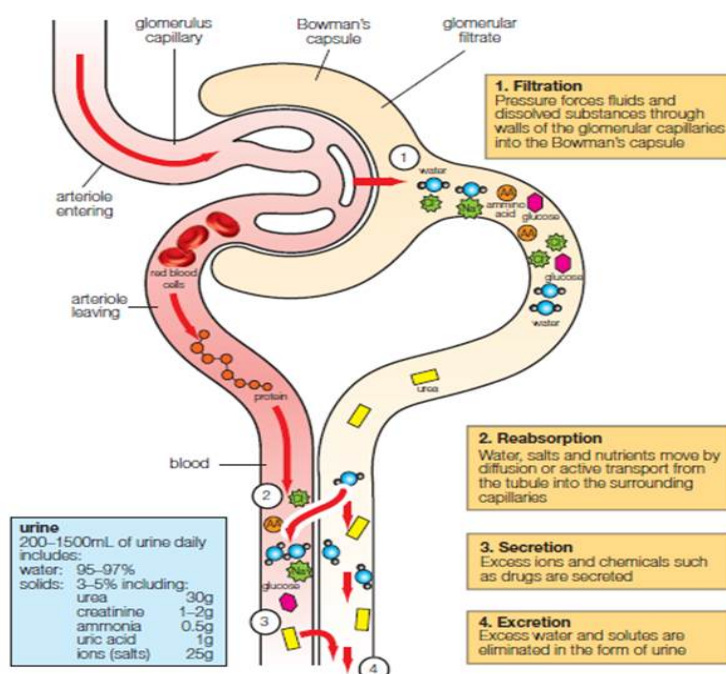
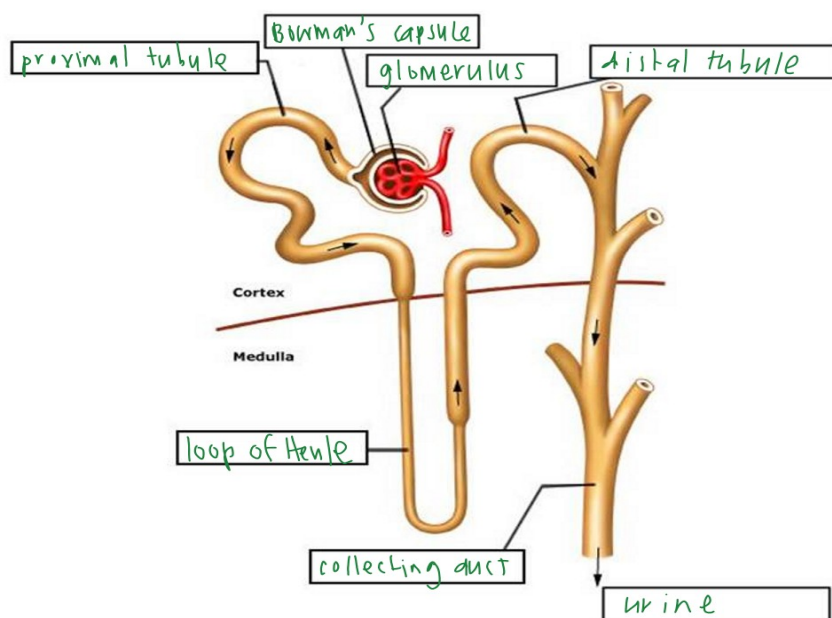
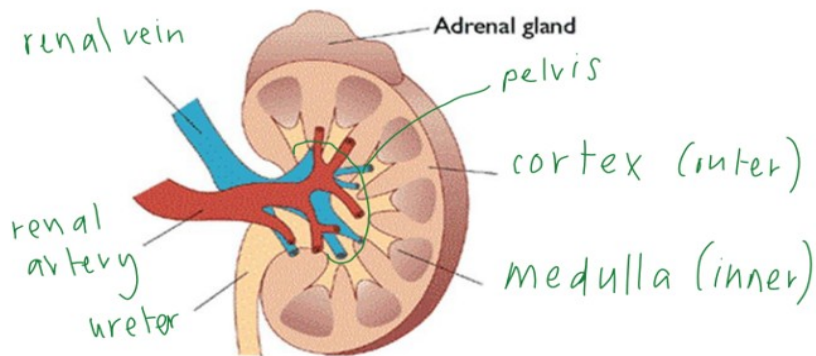


EYE DISORDERS + TECHNOLOGIES

- **Myopia (short-sightedness)** = eye can focus on near objects, but the focal length when viewing distant objects is too short
 - Causes:
 - Eyeball too elongated so image falls in front of retina
 - Lens cannot become thin enough to focus distant objects
 - Refractive power of cornea is too great
 - Technologies used to correct:
 - Concave lenses → spread out light rays to increase focal length
 - Laser eye surgery → changes curvature of cornea to decrease refractive power
- **Hyperopia (long-sightedness)** = eye can focus on distant objects, but the focal length when viewing near objects is too long
 - Causes:
 - Eyeball is too short so image falls behind retina
 - Lens cannot become round enough to focus close objects
 - Refractive power of cornea is inadequate
 - Technologies used to correct:
 - Convex lenses → converge light rays to shorten focal length
 - Laser eye surgery → changes curvature of cornea to increase refractive power
- **Cataracts** = any loss of transparency of the lens, causing blindness if untreated
 - Causes:
 - Ageing process
 - Congenital abnormalities
 - Disease e.g. **diabetes** or **glaucoma**
 - Technologies used to correct:
 - Phacoemulsification → probe emits EM waves that break up the lens, debris is removed and new artificial lens is inserted

- loss of kidney function: dialysis, transplantation

STRUCTURE OF KIDNEY



FUNCTION OF KIDNEYS

- The kidneys have two functions: osmoregulation + the removal of nitrogenous wastes (urea).
 - Wastes are converted into ammonia in the liver, however this is toxic so it is converted again into urea and excreted with water by the kidneys.
- Kidneys (mostly adrenal glands) hormonally control homeostasis by:
 - Activating vitamin D to release renin → raises blood pressure
 - Releasing erythropoietin → increases production of red blood cells
 - Releasing aldosterone → increases reabsorption of salts and thus water by osmosis
 - Releasing cortisol → controls salt and water balance

FUNCTION OF NEPHRON

Structure	Function
Nephron	• Functional unit of the kidney – regulates fluid composition by filtration, reabsorption, secretion, and excretion.
Renal artery	• Blood vessel entering nephron containing urea, glucose, salts etc.
Glomerulus	• Network of capillaries where blood components are squeezed under high pressure into the nephron (filtrate).
Bowman's capsule	• Filters blood from the capillaries based on SIZE – only small particles e.g. water and salts can enter the nephron.
Proximal tubule	• Useful substances e.g. water, glucose, amino acids are reabsorbed into the blood by active transport. • Bicarbonate ions reabsorbed to help regulate pH of surrounding blood.
Loop of Henle	• Water is absorbed in the descending loop and salts are reabsorbed in the ascending loop.
Distal tubule	• Selective reabsorption of ions and water occurs.
Renal vein	• Blood vessel leaving nephron containing filtered blood.
Collecting duct	• Has selectively permeable walls that allow for final reabsorption of water. • Urea is concentrated and combined to form urine.

PROCESS OF FILTRATION

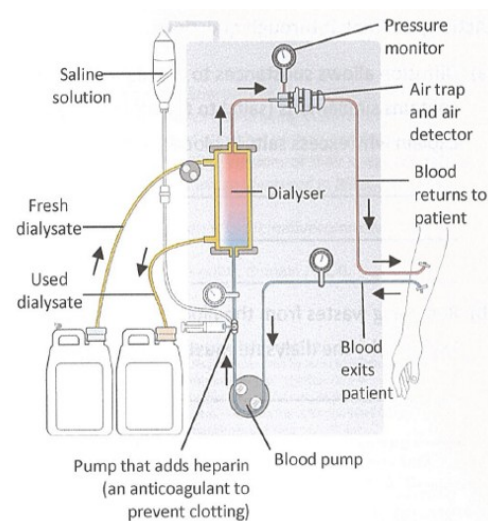
- Filtration in the glomerulus allows urea, water, vitamins, minerals, amino acids, and hormones to enter the nephron.
- The proximal tubule reabsorbs glucose, amino acids, bicarbonate ions and other nutrients.
- The descending part of the loop of Henle reabsorbs water.
- The ascending part of the loop of Henle reabsorbs salts.
- The distal tubule selectively reabsorbs water and mineral ions to control blood fluid concentrations.
- The collecting duct selectively reabsorbs water.
 - If blood water levels are low, the walls become permeable and water moves out by osmosis.
- Urine exits and travels to the bladder via the ureter to be excreted.

SUMMARY OF HORMONAL CONTROL

Too much water in blood?	Not enough water in blood?
Hypothalamus stimulates pituitary gland to release less ADH into blood.	Hypothalamus stimulates pituitary gland to release more ADH into blood.
Less water is reabsorbed into blood by the kidneys	ADH travels in blood to the kidneys where it causes more reabsorption of water into blood.
More water in urine	Less water in urine
Less water in blood	More water in blood

TECHNOLOGIES FOR KIDNEY FAILURE

- Kidney transplant → preferred treatment but difficult to access.
 - o Benefits:
 - Prolongs life
 - Offers a more active life, as no dialysis is needed
 - No restrictions on fluid or dietary intake
 - o Limitations:
 - Wait lists are long, as a low number of kidneys are available
 - Transplant organs must be carefully matched to prevent rejection
 - Patients with transplants must take immunosuppressants for the remainder of their lives to avoid rejection = more vulnerable to infection
- Dialysis → a life-prolonging treatment (renal replacement therapy).
 - o Process of haemodialysis:
 - Patient's blood is pumped through a dialyser
 - Blood passes through semi-permeable tubes surrounded by dialysate
 - Excess substances diffuse out of the blood, and filtered blood is returned to the patient
 - o Benefits:
 - Does not cause pain
 - Can increase lifespan and improve quality of life
 - Removes wastes and regulates fluids
 - o Limitations:
 - Many people still die from kidney failure while on dialysis
 - Very time consuming
 - Risk of infection at injection site



5.3 evaluate the effectiveness of a technology that is used to manage the effects of a disorder

TECHNOLOGIES

- Technologies may have adverse effects or require regular costs and maintenance.
- However, the side effects are outweighed by the benefits → can allow people with disorders to live a more normal life or prolong their life.

COCHLEAR IMPLANTS

- Benefits:
 - Allows deaf people to hear various sounds e.g. cars or alarms for survival
 - Can help children assimilate into mainstream schools and feel more comfortable
 - Allow people to learn spoken language and communicate → highly effective for children
- Limitations:
 - Frequencies are limited because of the few electrodes → cannot fully restore hearing
 - Require surgery, risking damage or post-operative infections
 - Transitioning to sound is difficult → less effective for adults

LASER SURGERY

- Benefits:
 - Quick procedure and quick recovery
 - Vision problems are fixed almost immediately
 - Usually no additional corrective eyewear is needed (9/10)
- Limitations:
 - Possibility of complications (1/10) e.g. glare, dry eyes
 - People with strong lens prescriptions will not have vision completely fixed
 - Expensive and risk of damage/infection

HAEMODIALYSIS

- Benefits:
 - Can occur in hospital or in home
 - Can be done during sleep
- Limitations:
 - Can take from 3-10 hours and needs to be done 3x per week
 - Requires external machine
 - Side effects include low blood pressure, vomiting, light-headedness, etc.

PERITONEAL DIALYSIS

- Benefits:
 - Uninterrupted time → most patients able to continue working
 - Can be done during sleep
- Limitations:
 - Need to be able to set up and attach equipment
 - Risks include hernias, weight gain, infection at catheter site, weakening of abdominal muscles, etc.