Module 8: non-infectious disease and disorders

Construct and interpret negative feedback loops that show homeostasis by using a range of sources, including but not limited to: (ACSBL101, ACSBL110, ACSBL111)

- temperature (ACSBL098)
- glucose

1.0 Homeostasis:

- Processes by which biological systems maintain stability and equilibrium by controlling different factors that play direct role for survival (temperature, pH, different chemicals etc).

Homeostasic mechanisms: nervous and endocrine systems maintain homeostasis

Differences	Nervous	Endocrine system
Form of signal	Electrical impulses (action potentials)	Chemical impulses
Pathway of communication	Transmission by neurons	Transmission in blood
Speed (relative)	Fast	Slow
Duration of effect	Short-term	Short or long-term
Type of action/response (voluntary vs involuntary)	Both	Involuntary
Target (localised or entire body)	Localised cells connected to a neuron	Many cells throughout body
Similarities		
Function	Regulation of body systems to maintain homeostasis	
Activation	Response to stimuli	

1.1 Negative Feedback Loops:

- Collection of processes that minimize/counteract the effect that causes disbalance in the normal body conditions.
- Mechanism involves opposing the condition that caused the normal body condition to deviate.
- Generally, a signal of change is sent to the control centre which in response, activates effectors which oppose the stimulus.

Detecting change	**Stimulus (change in external environment)**
	Sensory cells (interoceptors) detect change
Feedback loop	Control centre analyses messages and initiates actions
	Effectors (muscles, organs, glands)
Counteracting change	Response (action carried out to return to original state)

1. Stimulus (e.g. boy tapped on back)
2. Receptor activated (touch receptors simulated by information)
3. Sensory neuron (information sent from receptor to spinal cord)
4. Spinal cord (information sent to brain)
5. Brain (processes info in appropriate location)
6. Spinal cord (info from brain to motor neuron)
7. Motor neuron (information sent from spinal cord to effector organs)
8. Effector organ (information received by receptor organ e.g. muscle)
9. Response

1.1.1 internal coordination systems

Interceptors	
Thermoreceptors	Detect changes in internal temperature. Located in thermoregulatory centre (hypothalamus), detects blood temperature in the brain
Chemoreceptors	Detect changes in chemical concentrations. Located in certain blood vessels, detect pH and chemical levels (e.g. Co2, O2)
Osmoreceptors	Detect changes in osmotic pressure, located in hypothalamus. Osmotic pressure is determined by substance concentration dissolved in blood plasma. Causes body response through regulating water levels
Systems	
Somatic/voluntary nervous system	Allows conscious control of skeletal muscles. Contains 12 cranial nerves and 31 spinal nerves
Automatic/involuntary system	Controls automatic body functions (heart, smooth muscle/organs, glands). Divided into 'fight or flight' system and 'resting and digesting'
Parasympathetic system	'Rest and digest'. Conserves energy by slowing heart rate, relaxing digestive muscles, decreasing gland activity
Sympathetic system	'Fight or flight'. Increases energy by increasing heart rate, tightening digestive muscles, increasing gland activity

Action potentials
All or none principle
gland

1.1.2 Temperature Homeostasis by Negative Feedback Loop:

- Rise or fall in the temperature is detected by sensors that relay this message to the temperature regulation centre of the brain located in the hypothalamus.
- Responses may occur in the following ways:

Temperature Rise **Temperature Fall**

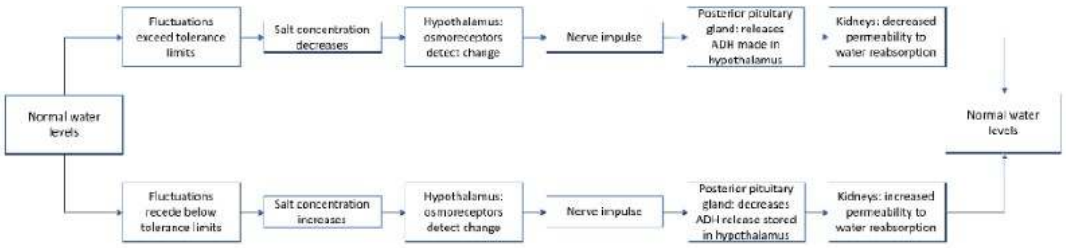
Vasodilation: Arterioles dilate (enlarge) so more blood enters skin capillaries and heat is lost.	Vasoconstriction: Arterioles get smaller to reduce blood going to skin: keeping core warm.
Sweating: Sudorific glands secrete sweat which removes heat when water changes state.	Shivering: Rapid contraction and relaxing of skeletal muscles. Heat produced by respiration.
Pilorelaxation: This means the hairs flatten.	Piloerection: Hairs on skin stand up.
Stretching Out: By opening up, the body was a larger surface area.	Curling Up: Making yourself smaller so smaller surface area.

1.1.3 Glucose Homeostasis by Negative Feedback Loop:

- Blood glucose level is controlled by hormones produced in the pancreas in the Islets of Langerhans.
- After intake of food, glucose level increases in blood which is detected by Beta Cells in the Islets of Langerhans.
- Beta Cells produce a hormone called Insulin.
- Insulin acts as a signal that triggers cells of the body, such as fat and muscle cells, to take up glucose for using as fuel.
- Insulin also encourages glycogenesis.
- If our body lacks glucose, Alpha cells produce Glucagon that convert glycogen to glucose and maintains blood sugar level.

1.1.4 water

Pituitary gland: produces antidiuretic hormone (ADH)



● investigate the various mechanisms used by organisms to maintain their internal environment within tolerance limits, including:
– trends and patterns in behavioural, structural and physiological adaptations in endotherms that assist in maintaining homeostasis (ACSBL099, ACSBL114)
– internal coordination systems that allow homeostasis to be maintained, including hormones and neural pathways (ACSBL112, ACSBL113, ACSBL114)
– mechanisms in plants that allow water balance to be maintained (ACSBL115)

1.2 Mechanisms to maintain internal environment

1.2.1 behavioural, structural and psychological adaptions in endotherms

Endotherms:

Their body metabolism generates heat

Metabolic processes maintain an internal body temperature that is independent of the external temperature.

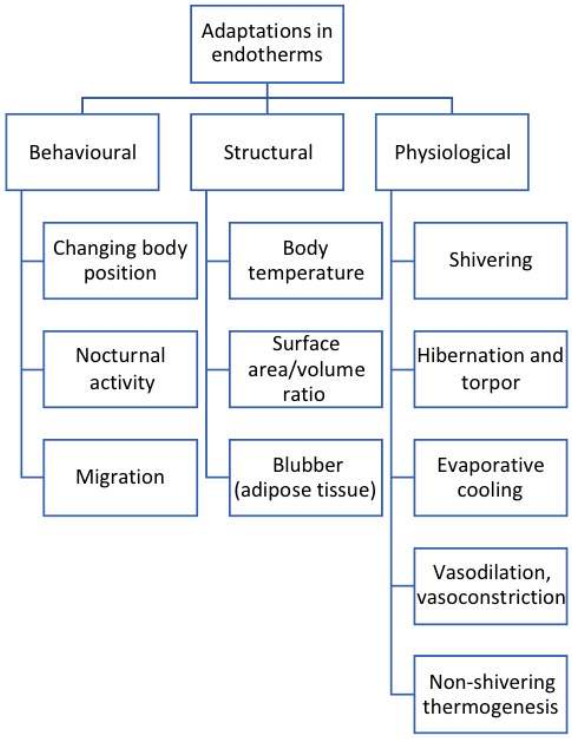
To do this requires energy, so more food is required by the endotherms.

Endotherm adaptions

Adaptations are features of organisms that aid their survival by allowing them to be better suited to their environment

These adaptations may be classified in a number of different ways:

- **Structural:** Physical differences in biological structure (e.g. neck length of a giraffe)
- **Behavioural:** Differences in patterns of activity (e.g. opossums feigning death when threatened)
- **Physiological:** Variations in detection and response by vital organs (e.g. homeothermy, colour perception)



Behavioural adaptions	
Body position (alter surface area)	Temperature too high – red kangaroo shades legs/tail with rest of body to reduce exposed area Temperature too low – mountain pygmy possum rolls into ball, reducing exposed surface area
Nocturnal activity	Typical where daytime temperatures are high Remain inactive during the day to avoid generating metabolic heat
Migration	Grey plover breeds in northern hemisphere in May-August and migrates to Australia August-April to avoid severe winter Humpback whales migrate annually from southern feeding grounds to warmer water to mate
Structural adaptions	
Temperature control	Includes insulation e.g. fur, hair, feathers to trap air to reduce heat loss
Body shape	Penguins have small ears and legs and round body shape. Minimises exposure to cold through small surface area/volume ratio Blubber (adipose tissue) prevents heat less through low thermal conductivity
Physiological adaptions	
Functions within the body	Metabolic activity (main source of internal heat)
Shivering	Increases metabolic rate, increasing internal body temperature
Hibernation	Extended period of inactivity in response to cold where heart rate and oxygen drop considerably but body temperature remains normal Mountain pygmy possum hibernates during cold winters to reduce energy expended on maintaining internal body heat
Torpor	Short-term hibernation where body temperature, metabolism, heart rate, and respiratory rate decrease Wombat slows its metabolism to a third of its normal rate on hot days *Hibernation and torpor are physiological rather than behavioural because of the changes in body function (i.e. Humans can't go into torpor)*
Evaporative cooling	Sweat
Vasodilation or vasoconstriction	Bilby ears regulate internal temperature through countercurrent exchange (warm arterial blood heats cooler blood in veins so organs are not cooled by returning blood from appendages)
Non-shivering thermogenesis below 36°	Fat in brown adipose tissue is broken down to produce heat, instead of usual carbs/sugars. Fats (lipids) are used for cellular metabolism, producing heat as a biproduct

1.2.2 internal coordination systems

Nervous and endocrine systems work separately or together to provide a message pathway via nerves or hormones from receptors to effectors.

Receptors

→ Detect stimuli (change from set point)

→ Are concentrated in sense organs eg eyes, skin

1.2.2.1 hormones

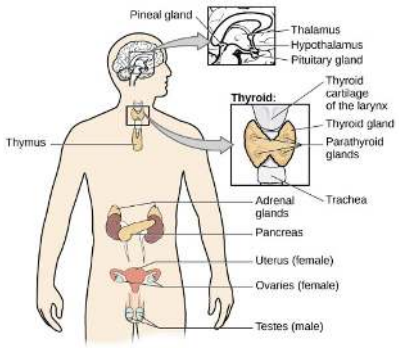
What is the endorcine system?

Ductless glads, release chemicals into the blood to regulate body functions

- Horomone is a chemical messanger that is transported via the blood stream to act on distant target cells
- Hormones are specific and will only activate cells or tissues that posses the appropriate target receptor
- Endocrine system is slower to initiate but has a mpre prolonged response when compared to the nervous system

Endocrine glands

- Secrete their product directly into the bloodstream, rather than through a duct
    - Major endocrine glands include, pancreas, adrenal gland, thyroid gland and the gonads
    - The hypothalmus and piturity gland and neuroendocrine glands an function to link the nervous and endocrine systems
    - Some organs may also secrete hormones despite not being glands



Examples of Endocrine Glands

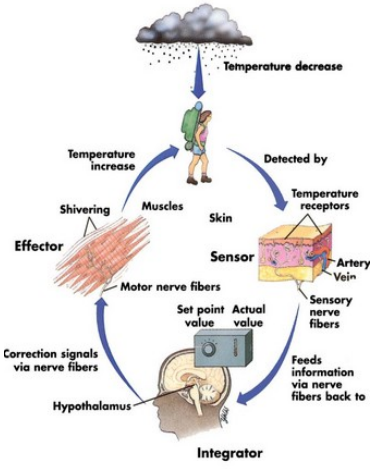
Gland	Hormone	Target Organ	Function
Pineal gland	melatonin	many	biological clock
Pituitary gland	FSH / LH ADH growth hormone oxytocin prolactin	ovaries kidneys many uterus breast tissue	menstrual cycle osmoregulation growth & division birth contractions milk production
Thyroid gland	thyroxin	liver	metabolic rate
Adrenal glands	adrenaline cortisol	many many	fight or flight anti-stress
Pancreas	insulin / glucagon	liver	blood sugar levels
Ovaries	estrogen / progesterone	uterus	menstrual cycle
Testes	testosterone	many	male characteristics

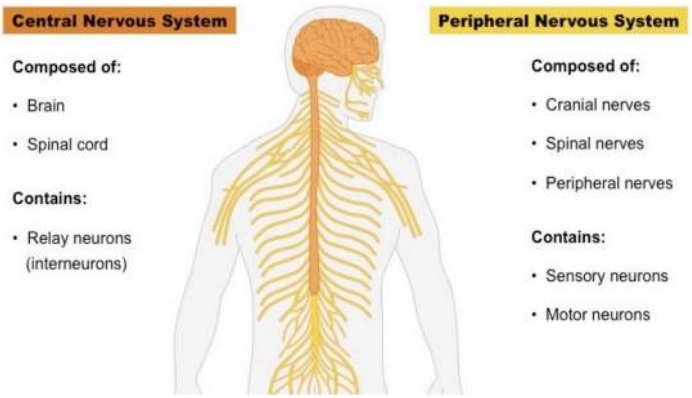
1.2.2.2 neural pathways

nervous system

- Coordinates actions of complex organisms via the transmission of electrochemical signals
    - The signals are transmitted by a specialised network of cells and neurons
- The nervous system divided into two parts
    - **Cns** - central nervous system- brain and spinal cord
    - **Pns** - peripheral nervous system- peripheral nerves, link CNS to body receptors & effectors

Main components of the nervous system





Part	Sensory Nerves	Effector	Receptor	Stimulus	Control Centre (CNS)	Response
Order	3rd	5th	2nd	1st	4th	6th

Neurons- function to transmit electrical impulses within the nervous system

- Converts sensory info into electrical impulses in order to rapidly detect and respond to stimuli

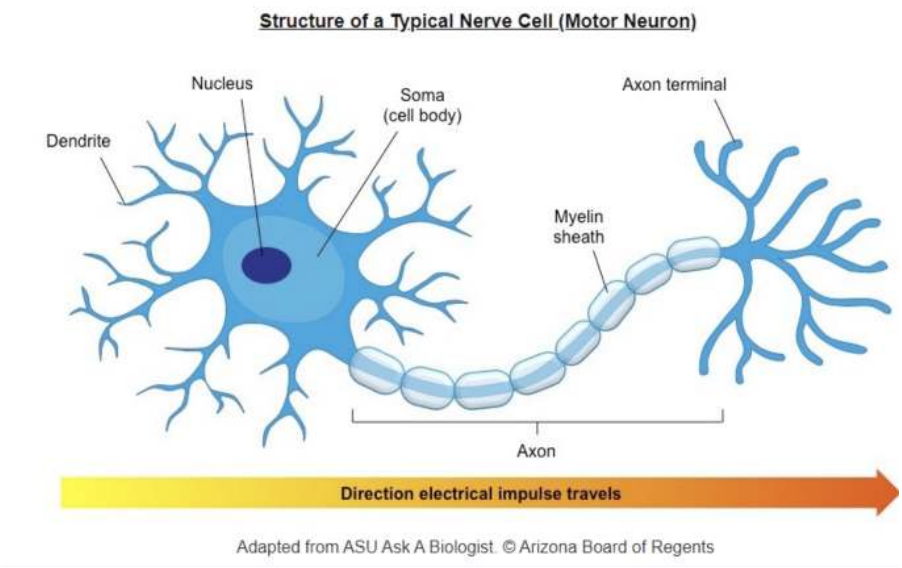
Neurons differ according to the role (sensory, relay or motor)

Three basic components

- **Dendrites-6** short branched fibres that convert chemical information from other neurons or receptor cells into electrical signals
- **Axon-** elongated fibre that transmits electrical signals to terminal regions for communication with other neurons or effectors
- **Soma-** a cell body containing the nucleus and organelles, where essential metabolic processes occur to maintain cell survival

In some neurons the axon may be surrounded by an insulating layer known as a myelin sheath

- Improves the conduction speed of electrical impulses along the axon, require additional space and energy

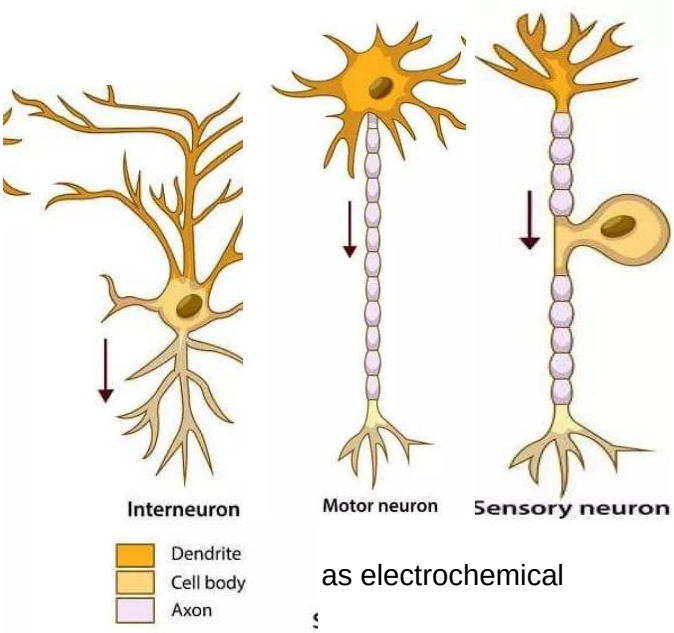


Types of neurons

Sensory - carry nerve impulses from sensory cells in PNS to the CNS.
(cell body on the side)

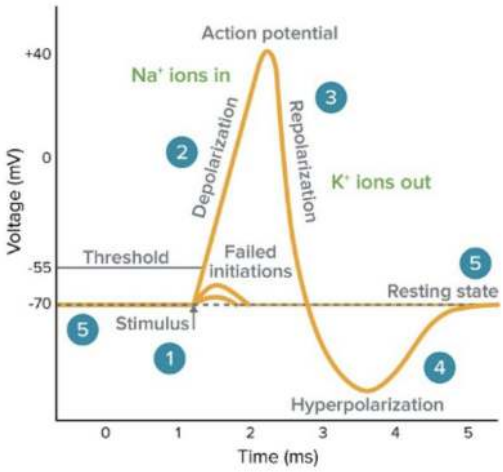
Motor - transfer messages from CNS to muscles or glands (effectors)

Interneurons - found within CNS and link sensory and motor neurons



How does a neuron work?

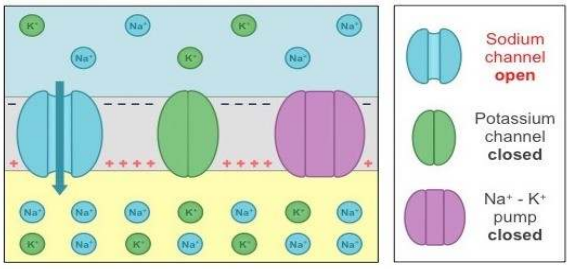
- ➔ Messages are carried by neurons as electrochemical impulses.
- ➔ There is a small gap called a synapse between the axon of one neuron and the dendrite of the next.
- ➔ The electrochemical impulse cannot travel over the gap and so a chemical called a neurotransmitter is released so the signal can keep moving.



action potential

When a stimulus is detected→ sodium channels in the cell membrane open up and sodium ions move into the neuron, reducing the potential from -70mv to -55mv.

This value is the threshold potential.



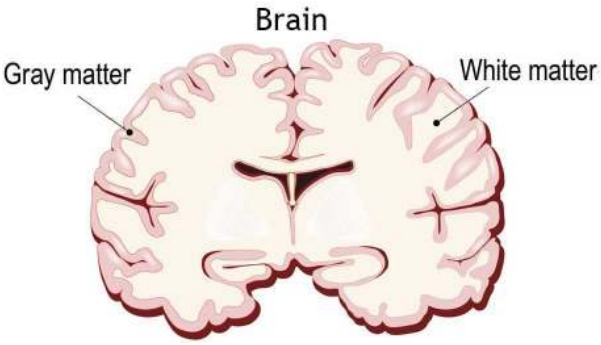
After: Na⁺ influx (*inside: +30 mV*)

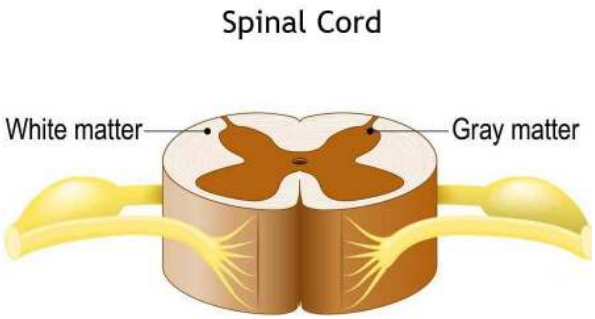
Central nervous system

The central nervous system (brain and spinal cord) can be characterised by two distinct regions – white matter and grey matter

- White matter is composed of bundles of myelinated axons which connect the various grey matter regions together
- Myelin acts as an insulator and hence nerve signals are transmitted at greater speed through white matter
- Grey matter is composed of the neuronal cell bodies and dendrites, as well as unmyelinated nerve fibres
- Grey matter functions as the regions of the brain where information is processed

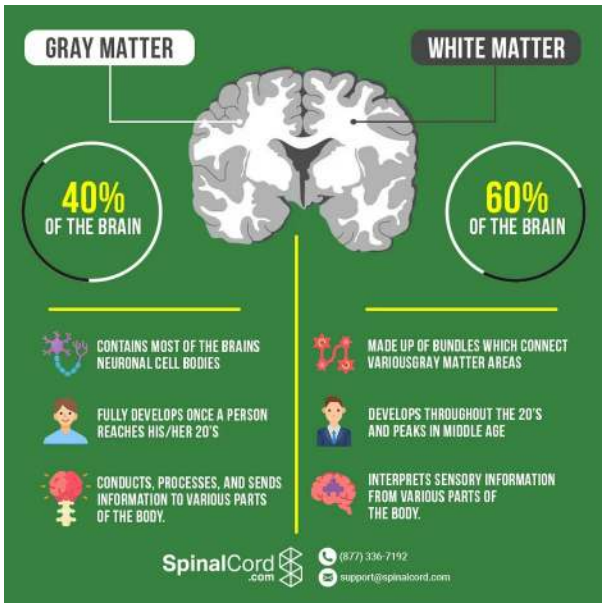
Brain	Spinal cord
Grey matter: neuron cell bodies **White matter:** nerve fibres + myelin sheaths Hypothalamus is control centre for homeostasis, maintaining heart rate, body temp, blood pressure, and O/CO blood concentrations	Conduction pathway for nerve impulses Coordinate reflex actions





Each region serves a different role.

- Gray matter is primarily responsible for processing and interpreting information.
- White matter transmits that information to other parts of the nervous system.



Hypothalamus

- Links the nervous system and endocrine systems in order to maintain homeostasis

- It receives information from nerves throughout the body and other parts of the brain and initiates endocrine responses
- It secretes certain neurochemicals (called releasing factors) into a portal system which stimulate or inhibit the pituitary gland
- It also secretes certain hormones directly into the bloodstream via neurosecretory cells that extend into the pituitary gland

  - pituitary gland lies adjacent to the hypothalamus and is in direct contact due to a portal blood system

- The pituitary gland is often referred to as the 'master gland', as it controls the secretion of a number of other endocrine glands
- The pituitary gland receives instructions from the hypothalamus and consists of two lobes (anterior and posterior lobe)
  - The anterior lobe (adenohypophysis) releases hormones in response to stimulation by hypothalamic releasing factors
  - The posterior lobe (neurohypophysis) releases hormones produced by the hypothalamus itself (via neurosecretory cells)

Spinal cord

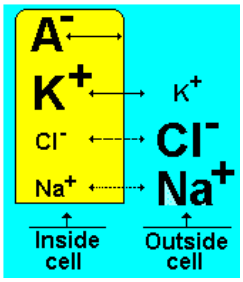
→ Extension of medulla oblongata

→ Acts as a pathway for nerve impulses from receptors to brain and then from brain to muscles and glands (effectors)

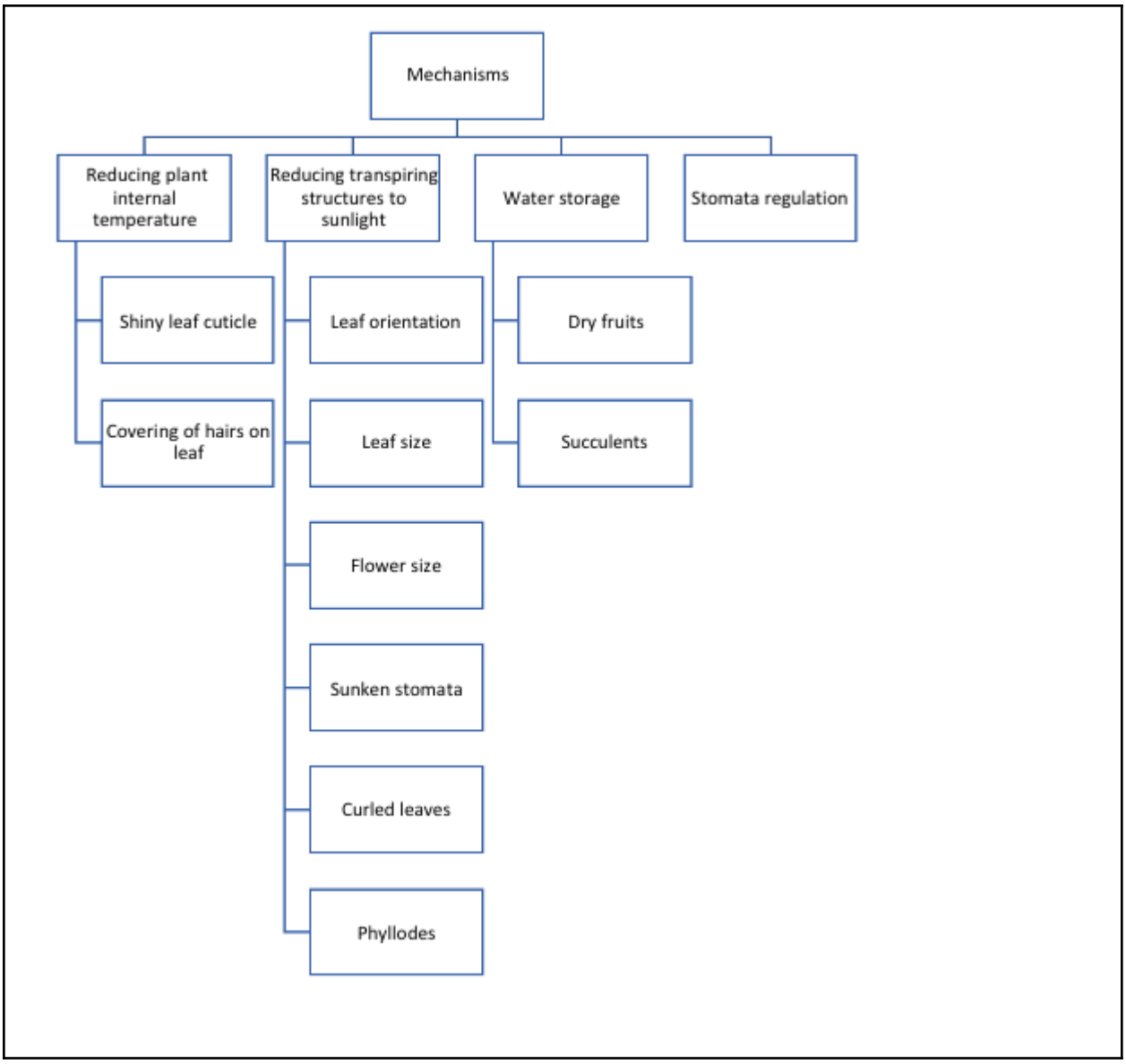
→ Also responsible for coordination of reflexes

Nerve impulse transmission

- ions on either side of the cell membrane of an axon.
- When there is no electrical impulse, there are more sodium ions outside the cell and more potassium ions inside the cell. The neuron is at rest.
- The resting potential of the neuron at this point is -70mv.



1.2.3 mechanisms maintaining water balance in plants

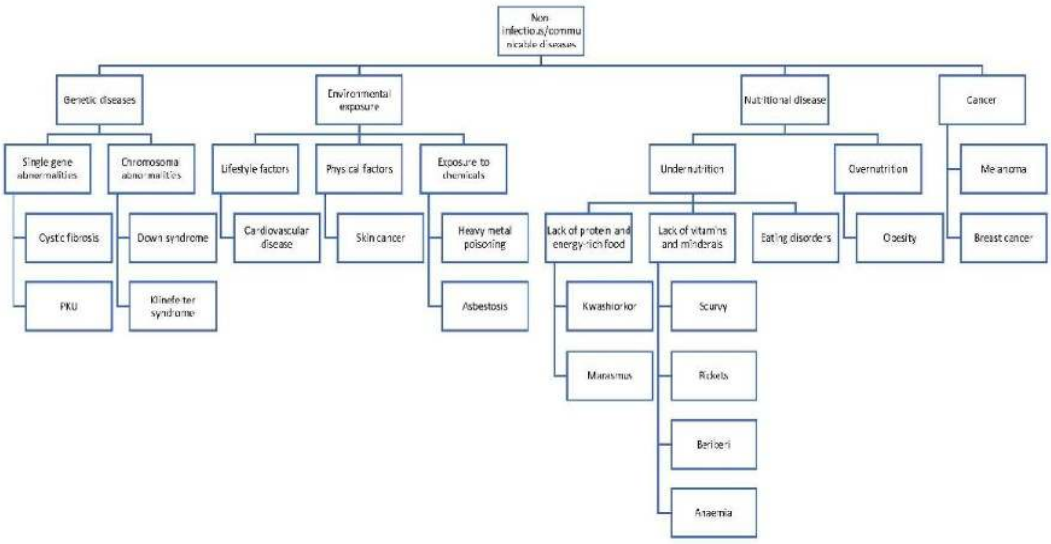

Mechanisms
Reducing plant internal temperature
Reducing transpiring structures to sunlight
Water storage
Stomata regulation
Shiny leaf cuticle
Covering of hairs on leaf
Leaf orientation
Leaf size
Flower size
Sunken stomata
Curled leaves
Phyllodes
Dry fruits
Succulents

- investigate the various mechanisms used by organisms to maintain their internal environment within tolerance limits, including:

– trends and patterns in behavioural, structural and physiological adaptations in endotherms that assist in maintaining homeostasis (ACSBL099, ACSBL114)

2.0 IQ: Do non-infectious diseases cause more deaths than infectious diseases

2.1 cause and effects of non infectious diseases in humans



2.1.1 Genetic diseases

Disease- any condition od the body or one od irs parts

- Impairs normal functioning and manifested by distinguishing signs and symptoms
- Genetic- abnormalities in genomes

Disorder- abnormal physical or menta;

Syndrome- symptoms that consistently occur together

Types of genetic diseases

2.1.1.2 Chromosomal abnormalities

- Chromosomal number mutations
- Structural chromosomal mutations
    - Translocations deletions or duplications of parts of a chromosome
- E.g. turner syndrome- aneuploidy called monosomy
    - Missing chromosome

	Causes	Effects
Down syndrome	Presence in zygote of additional chromosome 21 (trisomy 21) Affects 1 in 800 births	Characteristic facial features (small flattened skull, short flat-bridged nose, wide set, almond-shaped eyes, skin folds on eyes), protruding tongue, small, folded ears, intellectual disability, infertility, susceptibility to infection
Klinefelter syndrome	Trisomy involving sex chromosomes – males have XXY Individual chromosomes can be changed or mutated. Genes on deleted sections are lost	Lower testosterone and sterility Feminine fat distribution and breast growth. Young boys often have learning disabilities

2.1.1.1 Single gene disorders

- Change to a single gene
- Single base substitution, single base deletion or a single base addition
- Can involve many bases within a single gene
- Dominant or recessive
- E.g. huntingtons disease
    - Caused by a mutation in the IT15 gdnd on chromosome 4
    - Everyone has a region in the IT15 gene which contains a “genetic stutter”; s string of CAG repeats
    - More than 40 repeats= HD

Multifactorial diseases

- Combination of factors
- E.g. breast cancer
    - BRCA mutations involve mutations to the BRCA1/BRCA2 genes which are tumor supressor genes
    - Someone who has inherited a brca mutation will not necessarily get breast cancer
    - Increases risk 5x
    - Radiation and exposure to chemical mutagens

Can be a mutation of a gene or can be a mutation of a chromosome

2.1.2 Diseases caused by environmental exposure

Caused by the interaction of humans with their environment throughout their lives

2.1.2.1 lifestyle

Arise as a direct result of the way in which individuals lead their lives

	Causes	Effects
Cardiovascular disease	**Atherosclerosis:** hardening of the arteries. Occurs due to insufficient activity, alcohol consumption, stress, smoking, imbalanced diet. Occurs via deposition of lipids in inner arterial walls, causing artery walls to become rough and thickened	Hinders blood flow and increases blood pressure (hypertension). Deposited plaque reduces elasticity and blood flow, can cause cerebral haemorrhage. Occlusion can occur in blood vessels, causing cardiac arrest or heart failure **Ischaemic heart disease:** atherosclerosis of coronary arteries **Cerebrovascular disease:** atherosclerosis of cerebral arteries

2.1.2.2 physical

	Causes	Effects
Skin cancer	UV light causes abnormal cell division through dimerizing and oxidative mutations	**Forms basal cell carcinomas, squamous cell carcinomas, or malignant melanomas (most serious, causes satellite colonies). Development of skin cancer can be prevented through protection and maintenance of melanocytes (produce melanin to protect skin from UV rays)**

2.1.2.3 chemical exposure

	Causes	Effects
Heavy metal poisoning	Unsafe building practices (e.g. lead paint), unsafe waste disposal, air pollution etc.	Lead poisoning symptoms include headaches, irritability, reduced sensations, aggressive behaviour, abdominal pain, poor appetite, constipation, anaemia Causes loss of developmental skills, behaviour/attention problems, hearing loss, kidney damage, reduced IQ, slowed growth
Asbestosis	Asbestos associated with asbestosis, lung cancer, and mesothelioma Asbestosis occurs when fibres accumulate in narrow branches within lungs or migrate to pleural lining	Causes chronic cough, chest pain, restricted breathing. Risk of cancer or mesothelioma increases if individual also smokes

2.1.3 Nutritional diseases

- Deficiencies in particular vitamins, minerals or other nutrients
- Inadequate or excessive nutrition

2.1.3.1 undernutrition - lack of energy and protein rich food

	Causes	Effects
Kwashiorkor	Lack of protein causes excess fluid in body tissues, causing swelling under the skin (oedema)	Failure to grow, enlarged liver, hair changes, apathy, irritability, increased susceptibility to infectious diseases
Marasmus	Gross food deprivation causes lack of protein and energy intake	Loss of subcutaneous fat and susceptibility to infection causes death by starvation or heart attack

2.1.3.2 undernutriotion- lack of vitamins and minerals

	Causes	Effects
Scurvy	Deficiency of vitamin C (ascorbic acid)	Poor wound healing, joint pain, bleeding gums, bones that do not heal, spontaneous haemorrhaging. If left untreated, causes death
Rickets	Deficiency of vitamin D Exposure to sun initiates reactions to produce vitamin D	Children: defective calcification of bones, retardation of growth, and skeletal deformities e.g. bowed legs Adults: causes osteomalacia, which weakens bones and muscles. Also associated with osteoporosis
Beriberi	Deficiency of vitamin B1 Responsible for growth, carbohydrate and amino acid	Retarded growth, weakened heart muscle, loss of appetite, confusion,
	metabolism and function of heart, nerves, and muscles	nerve inflammation, poor coordination, tingling, and paralysis
Anaemia	Deficiency of iron Iron is essential component in haemoglobin, responsible for carrying oxygen around the body	Pale skin, weakness, unusual tiredness, apathy, low resistance to cold temperatures, difficulty breathing when exerted

2.1.3.3 eating disorders

	Causes	Effects
Anorexia nervosa	Characteristic features include psychological disorders, excessive weight loss, distorted body image. Developed fear of food and eating causing inadequate diet	Inadequate diet with reduced energy intake, excessive strenuous exercise, purging, appetite suppressants, diuretics, laxatives Effects include excessive weight loss, anaemia, impaired digestive function, bruising, low disease resistance, infertility, organ failure, death

2.1.3.4 overnutrition

	Causes	Effects
Obesity	Caused by consuming more kilojoules than energy expended, resulting in fat accumulation BMI used to measure total body fat. Used to determine whether the person is within the normal weight range, overweight, or obese - does not distinguish between fat and muscle (muscle is denser therefore heavier). Elite athletes with high muscle mass can display high BMI	Increased blood pressure, atherosclerosis, gallbladder disease, stroke, T2 diabetes, joint problems, increased risk of cancers e.g. colorectal cancer, reduced life expectancy

2.1.4Cancer

- A disease of the cells of the body
- Occurs as a result of abnormal cell division
- These cells can spread to the rest of the body and disrupt normal body functions
- Cell division is regulated by genes - DNA repair genes, proto-oncogenes and tumour suppressor genes

DNA repair genes - code for proteins that stop the cell cycle and can remove damaged sections of DNA and replace them with the correct sequence.

Mutations to DNA repair genes will result in cancer

Multifactorial disease

- Complex group of diseases with numerou contributing causes

Main causes

- Genetic predisposition
    - The chance of developing cancer is much greater if the person has a mutant copy of certain genes
    - Tumor supressor genes: protect a cell from one step on the path to cancer, p53
    - Protein encoded by the p53 gene has several anti-cancer actions
        - Activates DNA repair proteins when DNA has been damaged
        - Binds DNA and activates sveral genes that are important in the cell cycle and by doing so arrests the cell cyc;e at G1/S damaged cells
        - int=itiate apoptosis
    - Several mutations to the p53 gene have been linked to an increased risk of cancer
- Mutations due to:
    - Carcinogenic chemicals
    - Radiation
        - Mutation can be spontaneous or induced by chemical mutagens or radiation
        - If the mutation takes place in a tumor suppressor gene or an oncogene it can lead to the development of cancer
        - Proto-oncogenes arw involved in normal cell growth and division
        - More active than normal they may become cancer causing genes

- A failure in apoptosis
- An oncovirus

	Causes	Effects
Breast cancer	BRCA1 is a tumour suppressor gene found on chromosome 17, believed to be responsible for coding for proteins involved in repair of PTEN gene. BRCA1 mutations increase risk of breast cancer PTEN: tumour suppressor gene limiting cell division and encouraging cell death. Regulates cell cycle and prevents excessive proliferation	Mutation of BRCA1 causes non-production of proteins required for repairing damage to PTEN, causing lack of cell cycle control and runaway cell division as PTEN remains damaged
Melanoma	Uncontrollable skin cell division (melanocytes) Risk factors include UV exposure, fair complexion, severe sunburn, irregular-looking moles, chemical exposure, compromised immune system, older age, male gender	Initially confined to one area. If left untreated, increases thickness to spread to other skin layers. Melanoma spreads to nearby lymph nodes, lymph vessels, and skin. Then travel to organs e.g. brain, bones, liver. Interferes with correct body function to eventually cause death

Sickle cell disease

Sickle cell disease is a group of disorders that affects hemoglobin, the molecule in red blood cells that delivers oxygen to cells throughout the body. People with this disease have atypical hemoglobin molecules called hemoglobin S, which can distort red blood cells into a sickle, or crescent, shape.

Symptoms:

- Low number of red blood cells
- Repeated infection
- Pain (caused by rbc getting stuck in small blood vessels)which can deprive organs such as heart and kidneys of oxygen, leading to organ damage
- Red blood cells break down prematurely and this leads to anaemia (which then causes fatigue and shortness of breath)

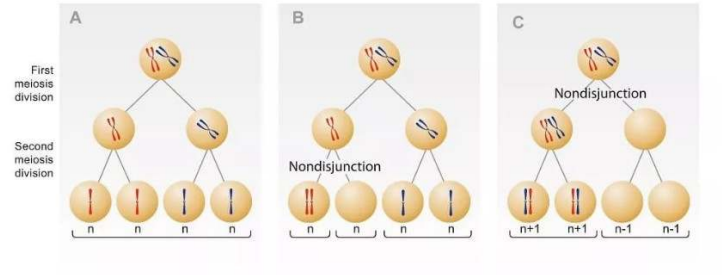
Frequency in a population

Sickle cell disease affects millions of people worldwide. It is most common among people whose ancestors come from Africa; Mediterranean countries such as Greece, Turkey, and Italy; the Arabian Peninsula; India; and Spanish-speaking regions in South America, Central America, and parts of the Caribbean.

Sickle cell disease is the most common inherited blood disorder in the United States, affecting an estimated 100,000 Americans. The disease is estimated to occur in 1 in 500 African Americans and 1 in 1,000 to 1,400 Hispanic Americans.

Chromosomal abnormalities

- Incorrect chromosome number (too many or not enough) by non-disjunction, trisomy or monosomy
- Deletion, addition or change in a chromosome



Nondisjunction during meiosis I and meiosis II

Down syndrome results when abnormal cell division involving chromosome 21 occurs. These cell division abnormalities result in an extra partial or full chromosome 21. This extra genetic material is responsible for the characteristic features and developmental problems of Down syndrome. Any one of three genetic variations can cause Down syndrome:

- **Trisomy 21.** About 95 percent of the time, Down syndrome is caused by trisomy 21 — the person has three copies of chromosome 21, instead of the usual two copies, in all cells. This is caused by abnormal cell division during the development of the sperm cell or the egg cell.
- **Mosaic Down syndrome.** In this rare form of Down syndrome, a person has only some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.
- **Translocation Down syndrome.** Down syndrome can also occur when a portion of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. These children have the usual two copies of chromosome 21, but they also have additional genetic material from chromosome 21 attached to another chromosome.

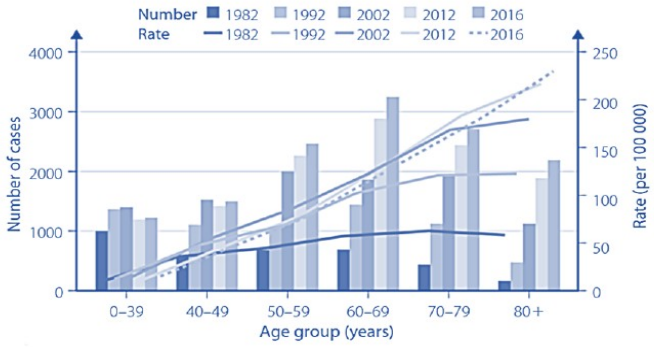
	Genetic	Environmental	Nutritional	Cancer
Disease name	Down syndrome	Mecury poisoining	Diabetes type 2	Skin cancer
Disease cause	trisomy 21 — the person has three copies of chromosome 21, instead of the usual two copies, in all cells	Inhaling mecury particles	obesity and an inactive lifestyle	Environmetal exposure to UV light
Effects on individual	Down's syndrome causes a distinct facial appearance, intellectual disability and developmental delays. It may be associated with thyroid or heart disease.	Organic mercury can damage your central nervous system (brain and spinal cord). Large amounts of mercury or long-term exposure can lead to death if not treated	doesn't produce enough insulin, or it resists insulin.	Causes mutations to the DNA of skin cells leading to abnormal cell division
treatment/ management		Stop exposure	Insulin injections diet	Surgery

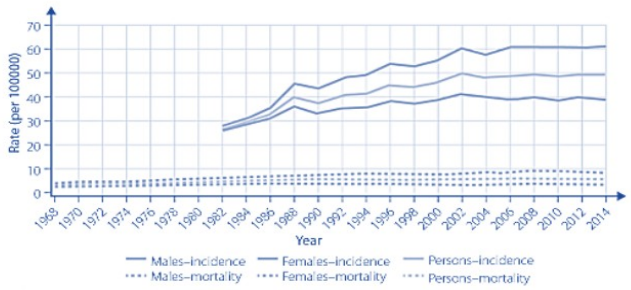
2.2 incidence prevelance and mortality rates of non-infectious diseases

Non-infectious diseases account for 70% global deaths/annum. 63% of these are due to lifestyle factors

- **Incidence:** number of new cases over specific period
- **Prevalence:** number of people with condition at specific time
- **Mortality:** number of deaths over specific period
- **Age-standardised rate:** measurement supposing standard age structure of population

2.2.1 diseases caused by environmental exposure





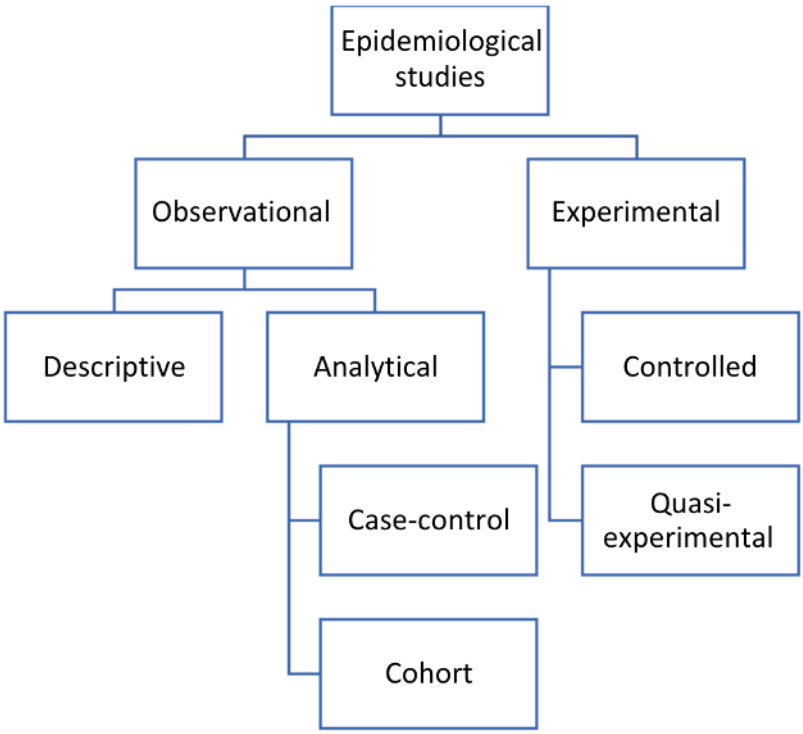
- Incidence and mortality rates of melanoma increase with age
- - Incidence rate of melanoma has steadied in Australia. Mortality is increasing. The rate of both is higher for males than females
- - Dark-skinned populations have lower incidence of melanoma than fair-skinned populations
- - Fair-skinned populations in higher latitudes (e.g. Australia, New Zealand) have highest global melanoma rates

- - Prevalence of melanoma is increasing and is greater in developed countries

8.3 IQ why are epidimiology studies used

Inquiry question: Why are epidemiological studies used?
● analyse patterns of non-infectious diseases in populations, including their incidence and prevalence, including but not limited to: ○ nutritional diseases ○ diseases caused by environmental exposure ● investigate the treatment/management, and possible future directions for further research, of a non-infectious disease using an example from one of the non-infectious diseases categories listed above ● evaluate the method used in an example of an epidemiological study ● evaluate, using examples, the benefits of engaging in an epidemiological study

3.0 epidemiology



Disease name	**description**	**type**	**Number of deaths in 2019**
Ischaemic heart disease	Collective term for diseases of the heart caused by narrowed heart arteries	environmental/ nutritional/ genetic	8.9 million
Stroke	a blood vessel that carries oxygen and nutrients to the brain is either blocked by a clot or bursts	Lifestyle genetic	6.2 million
Chronic obstructive pulmonary disease	a group of diseases that cause airflow blockage and breathing-related problems		3.1 million

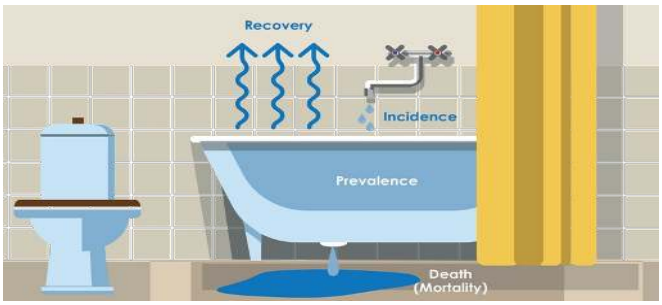
Lower respiratory disease	Infections of the lungs or below the voice box I.e: pneumonia, bronchitis, and tuberculosis.	Infectious	2.8 million
Neonatal conditional	disturbance of normal state of body, organs and abnormal function of a newborn		2 million
Trachea, bronchus, lung cancers	Cancers in the windpipes, lungs		1.9 million
Alzheimer's disease and other dementias	brain disorder that slowly destroys memory and thinking skills and, eventually, the ability to carry out the simplest tasks.	Brain non-infectious	1.8 million
Diarrhoeal disease	the passage of three or more loose or liquid stools per day		1.7 million
Diabetes mellitus	a disorder in which the body does not produce enough or respond normally to insulin, causing blood sugar (glucose) levels to be abnormally high.	nutritional/genetic	1.7 million
Kidney diseases	Longstanding disease of the kidneys leading to renal failure.The kidneys filter waste and excess fluid from the blood. As kidneys fail, waste builds up		1.6 million

Epidemiology:

- collect data relating to incidence, prevalence and mortality for both infectious and non-infectious diseases.
- used to determine possible causes of disease, risk factors and also possible methods of control of a disease.

Valid epidemiology studies:

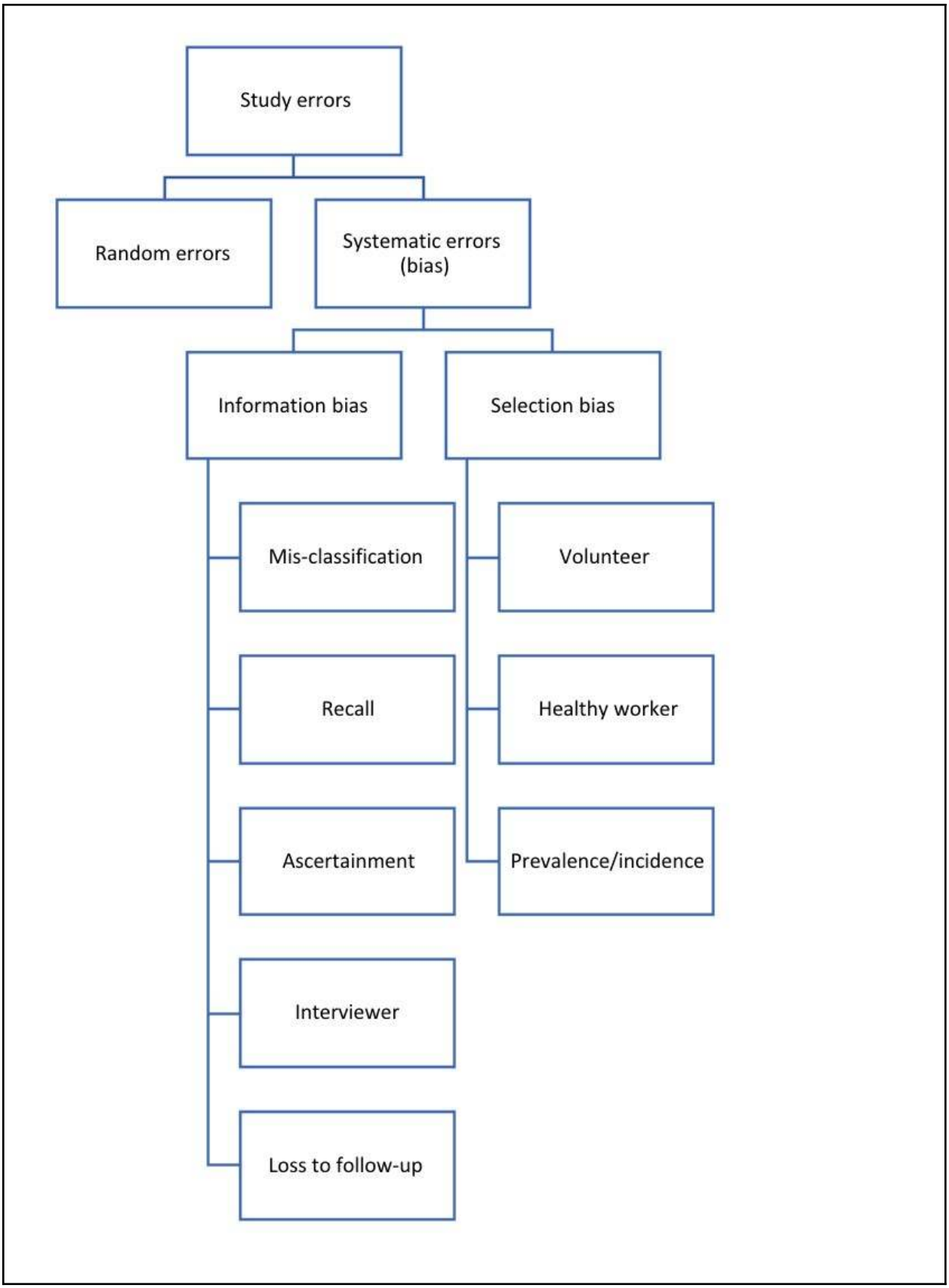
- Have large sample sizes
- Select populations with unequal exposure to possible causes
- Collect data on other factors which may affect disease like age, sex



Observational	
Descriptive	The first study conducted - Frequency - Segmentation of affected population - Location/time period Data collected from affected individuals, commonalities determined (e.g. age, sex, diet, occupation, socioeconomic status) Based on the data and commonalities, hypotheses are established for the potential causes
Analytical	Collect more date and statistically analyse to test hypotheses, examining: - Morbidity (no. cases past and present) - Mortality (death rate) - Prevalence (no. cases at a point in time) - Incidence (no. new cases over a period) Considers: - Factors preceding epidemic - Factors affecting risk of contraction - Control/match groups (similarities and differences in those with/without disease)

Case-control	Examine individuals with/without disease, hence importance of having a predetermined hypothesis. Includes - Differences in exposure (e.g. sunlight, asbestos) - Lifestyle factors (e.g. range of diets)
Cohort	Examine similar healthy individuals - Differences in levels of exposure to pathogens

Experimental Usually clinical trials for new drugs. Features matched controls, large groups, statistical analysis, conduced over a long period of time. Representative of broad range of society/lifestyle	
Controlled	Participants randomly placed into 2 groups. One receives drug and one receives placebo **Blind experiment:** doctors are unaware of the groups (reduces bias) Important to be done over a long period
Quasi-experimental	Impossible to randomise trail for whatever reason Researches choose who receives drug


Study errors
Random errors
Systematic errors (bias)
Information bias
Selection bias
Mis-classification
Recall
Ascertainment
Interviewer
Loss to follow-up
Volunteer
Healthy worker
Prevalence/incidence

Random errors	
Unpredictable variations	Inconsistent effect on measurement Corrected using statistics Decreased precision (does not skew results) Can occur through differences in subjects
Systematic errors (bias)	
Information bias	Errors in taking measurements/recording information
Misclassification	Patient suffering from condition but asymptomatic – put into the wrong category
Recall	Those affected have a much higher recall
Ascertainment	Not followed up to the same degree
Interviewer	Interviewer bias, asks leading question
Loss to follow-up	People are unavailable at the end of the study
Selection bias	Errors occurring due to subject selection/sampling (how/why/where subjects are chosen)
Volunteer	Vested interest/genetic link
Healthy worker	Healthier subjects tend to be used (not representatives)
Prevalence/incidence	Failure to include all cases (e.g. recovered, died)

3.1 analysing patterns of non- infectious diseases

- Identify patterns in the incidence, distribution prevalence and mortality rates of disease
- Possible cause of disease and whether certain population groups are at greater risk of developing a certain disease
    - Develop strategies that would be most effective for controlling the disease in a population

- Scientific and mathematic models used to statistically analyse the data that has been collected, to provide information about the trends
- Determines trends
- Gender, race and location
- Tables and graphs

Incidence	New cases over specific period (diagnosed or reported)
Prevalence	Total cases over a period OR number of people still alive
Mortality	Deaths over a specific period
Age-standardised rate	What the rate would be given a standard age structure – weighted mean of age-specific rates (to compare populations)

3.2 Treatment and management for one example of non-infectious disease (melanoma and diabetes)

Difference between <u>treatment</u> and <u>management</u>

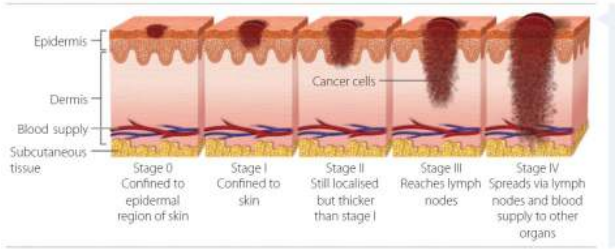
<u>Treatment</u>- depends on type of disease

- Eg. vitamin and mineral intake correction to treat vitamin and mineral deficiencies
    - Scrutvy- more vitamin c
- Giving them the necessary medicines and therapy to cure them biologically
- Only physical aspect of the illness and includes methods to cure them

<u>Management-</u> how to deal with the disease

- Managing symptoms
- treat them not only physically, but also giving them mental and emotional care
- Addressing them about their cure, personal chates

Melanoma- different treatments for different stages



Melanoma treatment-

Treatment options: https://www.youtube.com/watch?v=bjN3XlTXaIc

Early detection- margin, surgery, often the only treatment

- Sentinel lymp nodes biopsy- swelling and lymphedema (skin redness or stiffness)
- Involves removing the tumor and the skin around the tumor, no cancerous cells left behind, reduce the risk of a recurrence of melanoma

Immunotherapy- stimulates the immune system

- Checkpoint inhibitors, t cells to attack cancer, block proteins
- Flu like symptoms, dirraheo

Targeted therapy- precision medicine

- Tumor may have changes to gene
- Chills, fever, itching, rass and extreme sensitivity

Radiation therapy-

- Swelling, body aches, skin burning
- Risk of getting second cancer

Cancer vaccines-

- Recognise and kill cancer cells
- Being tested for melanoma-not yet exist

3.2.1 future directions of treatment

- Further develop targeted therapies and immunotherapy treatments for melanoma
- Targeted therapies is based on the different types of mutations that cause the uncontrolled cell division
- Interrupt specific pathways
- Further research required to develop a greater variety of immunotherapy drugs so that the majority of patients can benefit from this form of treatment of melanoma

- Use of vaccines to treat melanoma is in it's early
- futher research may also involve investigating the relatiolnship between melanoma and other cancers

3.3 Evaluation of method used in epidemiological study

3.3.1 what is an epidemiological study

How often a disease occurs in different groups of people and why

3.3.2 evaluating the epidemiological study of skin cancer

Skin cancer and outdoor occupations-

Adèle green, et al	Hypothesis that the prevalence of skin cancers in outdoor workers
	Found no significant correlation between occupationa and incidence of skin cancer
At nambour queensland	Nambour has a very high prevalence of skin cancers 3,00 nambour residents chosen at random from the electoral roll, all invited to participate of which 70% accepted the invitation

flaws	- People who choose to participate are more likely to be
	  - Unemployed or retired or have time to spare
	  - Socially orientated and/or concerned about skin cancer

3.4 Evaluation of benefits of epidemiological study

Making a judgement about something and using evidence to support your judgement.
Evidence should be based on specific criteria relevant to the particular scenario being evaluated
Validity of the method used in a epidemiological study should be evaluated on hether it follows accepted epidemiological principles
Any errors should be considered when assessing the validity of the study

KEY CONCEPTS

- To 'evaluate' means to make a judgement about something, based on certain criteria. Evidence should be provided to support the judgement that has been made.
- Evaluation of the validity of the method used in an epidemiological study is determined by how well it follows accepted epidemiological principles.
- Along with large sample size and long periods of study, the use of scientifically approved methods of implementing a study, collecting data and analysis of results are the main requirements of epidemiological studies.

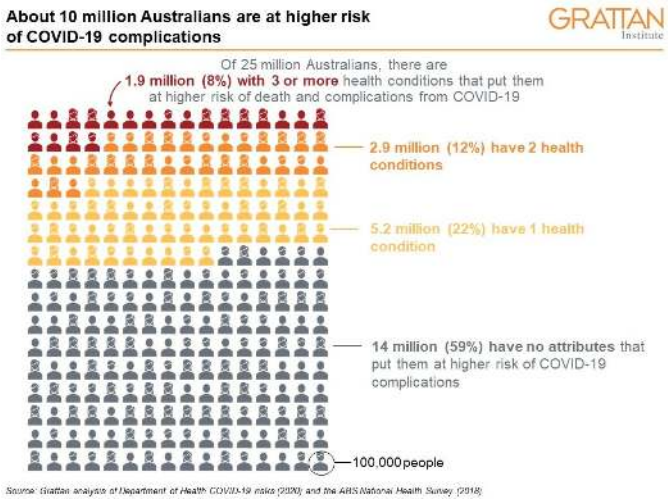
3.4.1 benefits of epidemiological study

- Benchmark the prevelanceof a disease and **monitoring** any changes over time
- Supply public health warning and **community education**
- Inform **public policy** and law-making

8.4 IQ how can non-infectious diseases be prevented

4.0 prevention

- Prevention is always better than treatment
- Reduces pain and suffering to an individual, reduces their burden on the health care system
- Reduces associated financial burden as well
- 1 in 2 aussies have a chronic disease
    - Cvd
    - Cancer
    - Diabetes
- Almost ⅓ can be prevented



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4.1 Effectiveness of current disease-prevention methods

- Data obtained from epidemiological studies identifies the diseases that are most prevelant in populations and the groups that are most at risk
- Assists health authorities and governments to develop strats aimed at preventing the diseases that are of most concern becuase of their incidence, prevelance and mortality

4.1.1 educational programs and campaigns

- Strategies to provide info and educate the population about the effects of a disease and the risk factors that increase the chance of developing that disease
- Hwo to avoid the risk factors, change behaviour to reduce their exposure to these risk factors and lower their chances of developing the disease

Features of a successful public health campaign:

- Evidence base for action
- Limited number of high priority evidence based interventions
- Effective performance management with real-time monitoring evaluatqion and program improvement
- Partnerships between the public and private sectors
- Communication of accurate info to health care workers and the public
- Political commitment

Components of public health campaigns:

- National days or weeks
- Online resources
- Advertising campaigns
- Social media
- Apps
- Posters
- Target audience
- Screening those at risk
- National help lines
- Health advice
- Legislation
- Funding for organisations that provide support services
- Price increases
    - Cigarettes
    - Alcohol

Current effectiveness of PKU prevention

- In aus- all babies born are tested for PKU
- Heel prick- to take blood and collected on a blotting paper
- Blood is placed on a special growth medium that conatinsn a growth inhibitor
- The bacteria bacillus subtilis cannot grow this medium, but high levels of phenylalanine can overcome the inhibition, allowing bacteria to grow - indicating PKU
- Babies who are identified as having PKU are immediately placed on a special diet
    - Low protein and other foods containing phenylalanine
    - Diet followed- normal, healthy, long life

4.1.2 genetic engeneering

Pre- implantation genetic testing

- The PAH gene which encodes the enzyme phenylalanine hydroxylase is located on the long arm of chromosome 12
- A genetic test is available for this gene locus

Two genetic tests

- Parents can be tested you d\etermine their carrier status
- An embryo can be testested to determine whether or not it has PKU

4.1.3 gene therapy

Introducing normal alleles into cells in place of missing or defective alleles into cells in place of missing or defective alleles in order to correct a genetic disease

- For PKU involve a normal allele of the PAH gene into the liver cells of a person with PKU
- PKU is recessive, sufferers are homozygous
- If a normal allele of the PAH gene is introduced into a scell, that cell will express phenylalinine hyfroxylase and be able to convert phenylalanine to tyrosine
- Several vectors

8.5 IQ technologies and disorders

5.1 structures and functions of organs

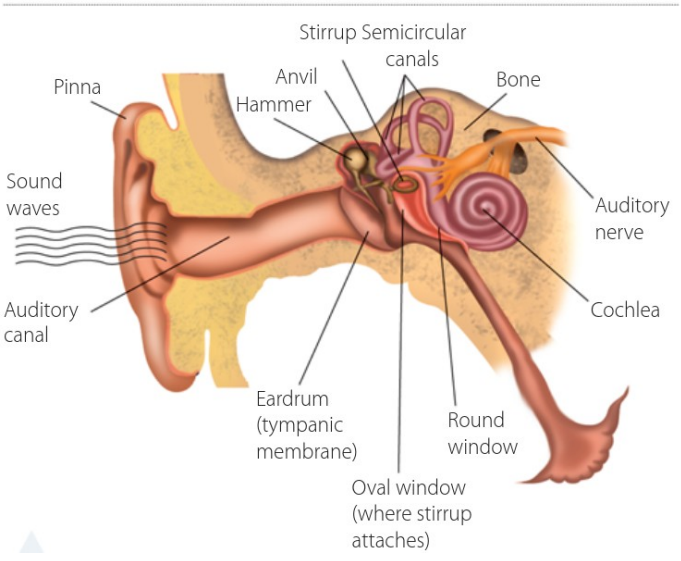
5.1.1 ears

Pathway of sound

External auditory canal→tympanic membrane→ossicles→oval window→cochlea→round window

The ear is a sense organ that provides a major communication pathway between the external environment and the body

- Problems with the structures and functioning of the ear can cause hearing
- The pathway of the sound wave through the ear can be summarised as:
    - Pinna→external auditory canal→ tympanic membrane→hammer, anvil, stirrup→ oval window→cochlea→ round window
- The organ of corti contains hair cells that are the receptors in the ear
- Bending of the hair cells when a pressure wave pushes on the basilar membrane stimulates the formation of electrical impulses
- Electrical impulses are transferred to the brain by the auditory nerve
- Conductive hearing loss occurs when vibrations cannot be transferred effectively through the outer and middle ear
    - Middle ear infection
    - Perforated eardrum
    - Damage to the middle ear bones
    - A benign growth in the middle ear
- Sensorineural hearing loss occurs when the inner ear is damaged or malformed
    - Exposure to loud noise
    - Age
    - Illness such as measles, mumps and meningitis
    - Genetic disorder



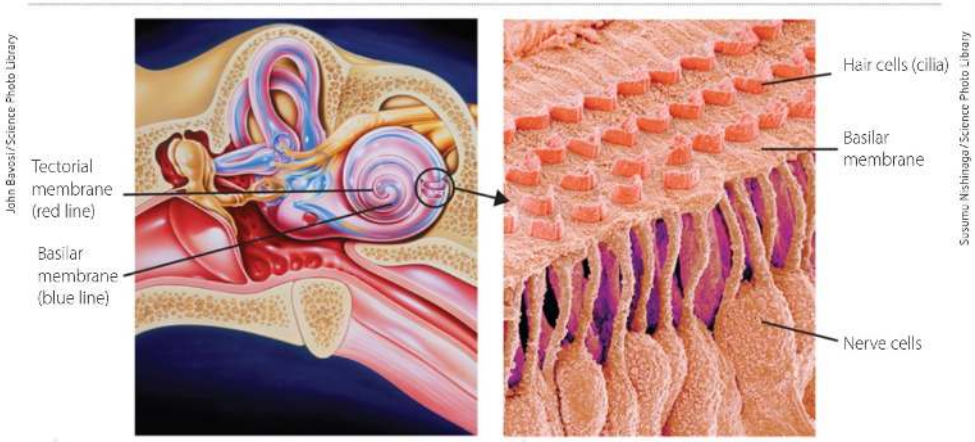
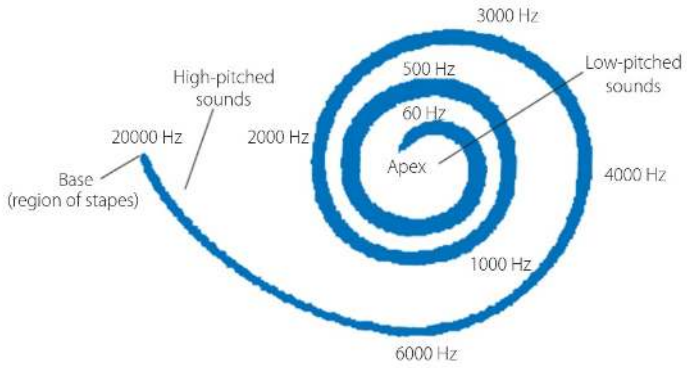


FIGURE 18.3 The inner ear consists of fluid-filled passages made up of three canals: vestibular membrane, tectorial membrane and basilar membrane

FIGURE 18.4 An SEM of hair cells within the cochlea showing the cilia, which bend with sound triggering a neurochemical response that generates nerve impulses.

FIGURE 18.5 Vibrations of different frequencies stimulate hair cells at different positions along the cochlea.



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5.1.2 eyes

- Rods and cones are the receptor cells in the retina of the eye. The visual pigments present in each of these cells change light energy into electrothermal impulses to be sent to the brain for processing
- Rods contain rhodopsin and are responsible for the detection of light and vision in low light
- Each cone contains one of three iodopsin pigments (blue, green and red. Cones are responsible for coulour vision. The multitude of colours detected is due to the combination of eac of the cones
- All pigments are composed of retinal (a derivative of vitamin a) and opsin (a proteom) the type of sopsin depends on the type of pigment
- The detection of light by the pigments stimulates the formation of an alectrothermal impulse and the splitting of the retinal and opsin, when the two parts of the molecule recombine the pigment is ready to receive more light energy
- Rods are distributed all over the retina
- Cones are in clumps with fewer on the edges of the retina
- Common visual disorders such as myopia and hyperopia are caused by refractive errors of the eye

- Cataracts are caused by clouding of the lens
- Macular degeneration of the cells in the layer beneath the retina

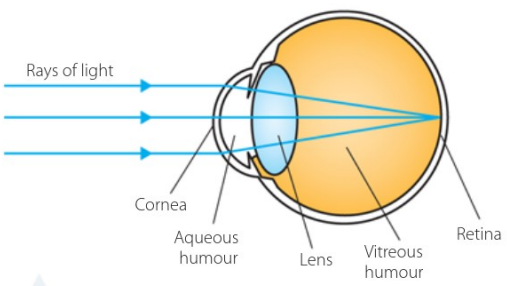


FIGURE 18.8 Refraction of light rays as they pass through the eye to form a sharp focus on the retina

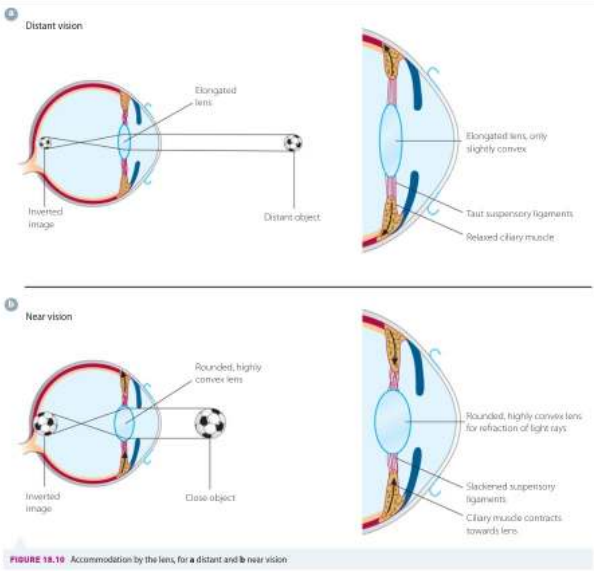


FIGURE 18.10 Accommodation by the lens, for **a** distant and **b** near vision

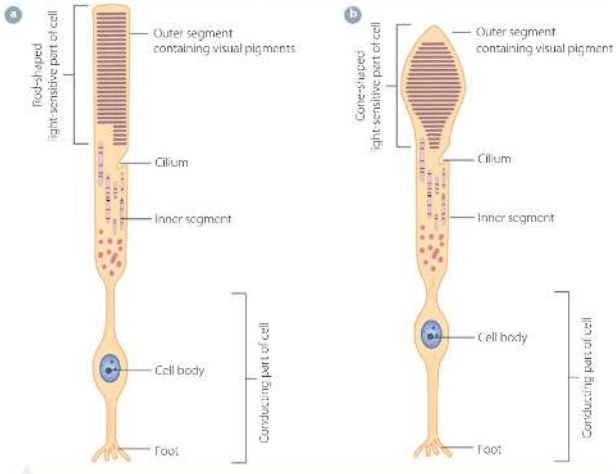


FIGURE 18.12 The structure of **a** a rod cell and **b** a cone cell

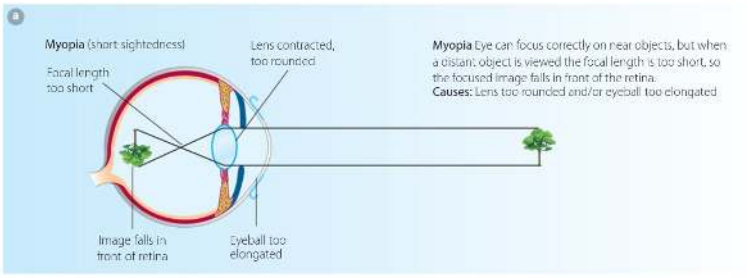


FIGURE 18.14 Two common visual disorders: **a** myopia (short-sightedness) and **b** hyperopia (long-sightedness)

Cataracts

A cataract is the clouding of the lens (Fig. 18.15), which reduces the transmission of the light through the lens. This causes blurred vision of both near and far objects, and increased sensitivity to the glare of bright sunlight.

FIGURE 18.15 Cataracts cause visual disorders due to clouding of the lens: **a** a normal lens; **b** a lens with cataract

5.1.3 kidneys

- Loss of kidney function can occur for numerous reasons, including diabetes, high blood pressure, kidney infections and blockages
- Most of these conditions cause damage to the fragile nephrons
- Dialysis uses and ‘artificial kidney’ to tremove ureaj from the blood when kidney function is insufficient to do so
- Dialysis can also balance the water and salt levels to come extent
- There are two types of dialysis: haemodialysis and peritoneal dialysis
- Renal dialysis patients undergo dialysis several times a week for extended periods
- The effectiveness of a technology can be assessed on the basos of certain criteriea

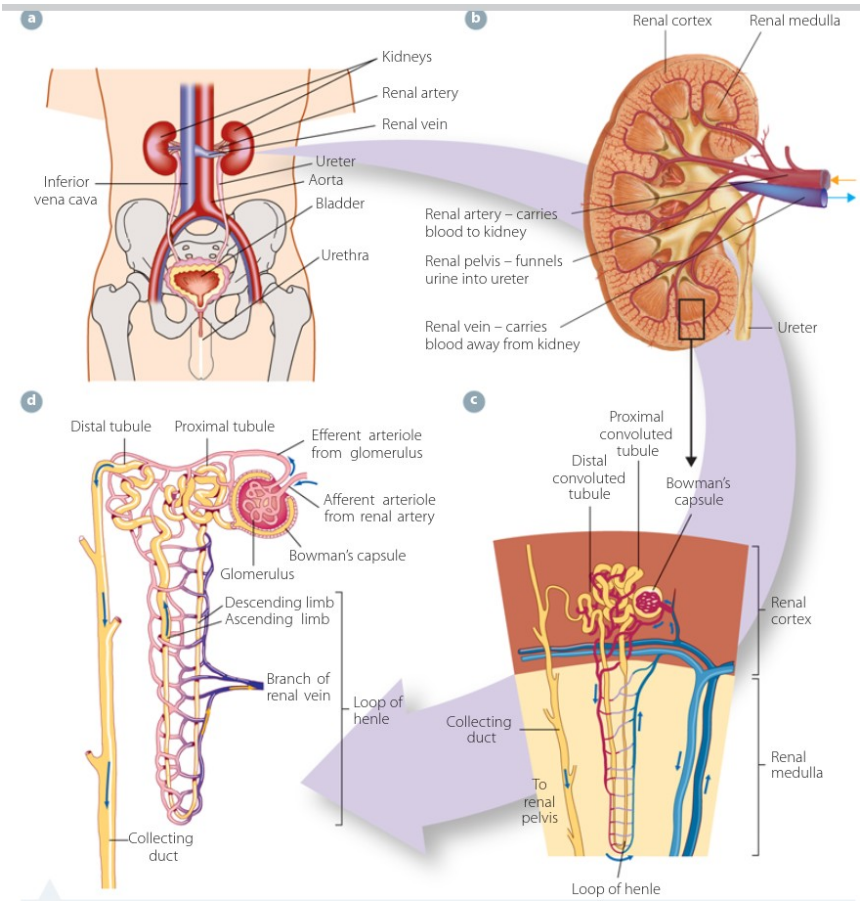


FIGURE 18.19 **a** Excretory system of mammals; **b** macroscopic structure of mammalian kidney (longitudinal section); **c** microscopic structure showing the distribution of tubules in the mammalian kidney; **d** nephron and associated capillaries

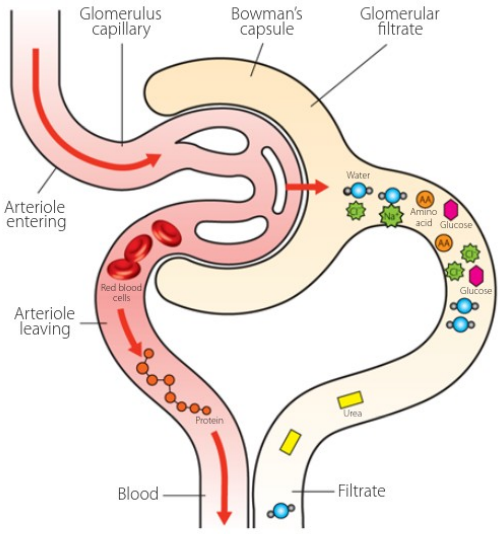


FIGURE 18.20 Filtration of blood in the Bowman's capsule

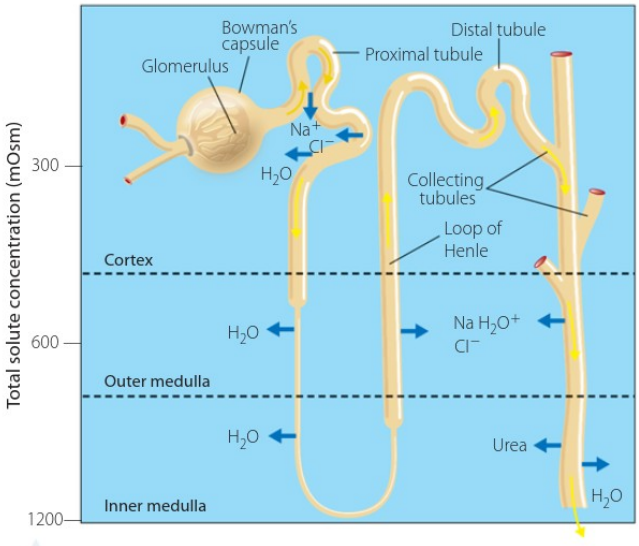


FIGURE 18.21 Reabsorption of water and various ions along the tubule. Blood vessels are not shown.

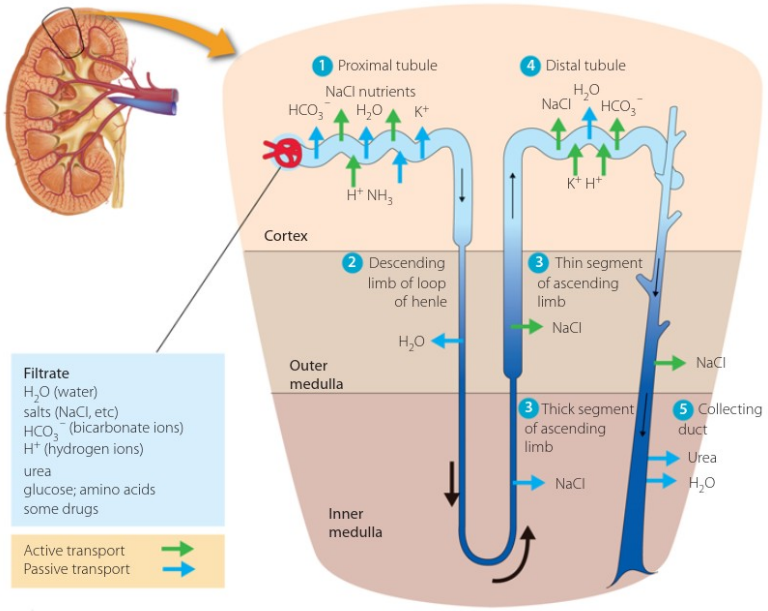
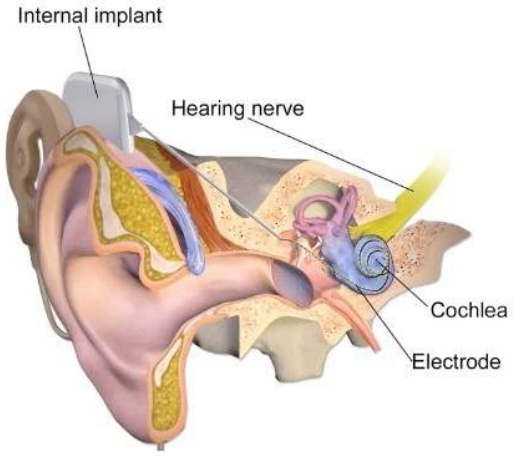


FIGURE 18.22 Reabsorption of ions and water from the tubules by active transport (ions) and passive transport (water). Blood vessels are not shown.

5.2 technologies used for disorders

5.2.1 Hearing loss: cochlear implants, bone conduction implants, hearing aids



Cochlear implants

- Electronic devices that are used to give hearing to those who are profoundly deaf
- Two parts
    - Outer part contains a microphone while the inner part
    - Includes an electrode that feeds into the cochlea and directly stimulates the hearing nerves

Bone conduction implants

- Used to improve hearing for those who have conductive hearing loss
- Transmit vibrations through bone to the inner ear, bypassing the defective outer and middle ear

Hearing aids

- Traditional hearing aids come in many different shapes and sizes and are fitted to a patient by an audiologist depending on the type and severity of the patients hearing loss
- Typically hearing aids help a person to hearing aids healp a person to hear by making sounds louder as they enter the outer ear

KEY CONCEPTS

- In cases of conductive hearing loss, hearing can be assisted by the use of hearing aids and bone conduction implants.
- Hearing aids magnify the sound waves to assist their passage through the outer and middle ear to reach the inner ear.
- Bone conduction implants bypass the outer and middle ear by sending the vibrations through the bone above the ear straight to the inner ear.
- Hearing aids and cochlear implants assist hearing in cases of sensorineural hearing loss.
- Cochlear implants involve the conversion of sound into electrical impulses that stimulate an electrode array implanted in the cochlea, which in turn stimulates electrochemical impulses in the auditory nerve.

5.2.2 Visual disorder: spectacles, laser surgery/intraocular lens implantation, bionic eye

Spectacles and contact lenses

- Correct the vision of people with hyperopia, myopia and or astigmatism, by refracting the light entering the eye in a way that is complementary to the defect in the eye

Laser eye surgery

- A small circular flap is created int eh cornea using a precise metal blade or a laser that creates a series of tiny closely- positioned bubbles in the cornea
- The flap is then folded back to reveal a layer of the cornea called the stroma

- A laser is then used to vaporise material in the corneal stroma, in order to reshape the cornea to correct for the refractive error in the eye
- Finally the flap is replaced and the eye allowed to heal

KEY CONCEPTS

- Lenses are used in spectacles and contact lenses to correct the refractive errors of the eye.
- Concave lenses correct myopia, while convex lenses correct hyperopia.
- Laser surgery changes the convexity of the cornea to correct refractive errors and allow light rays to be focused on the retina.
- Cataract and laser cataract surgery restore vision to cataract sufferers.
- The bionic eye could help to restore vision to those suffering from retinitis pigmentosa by bypassing damaged rod and cone cells.

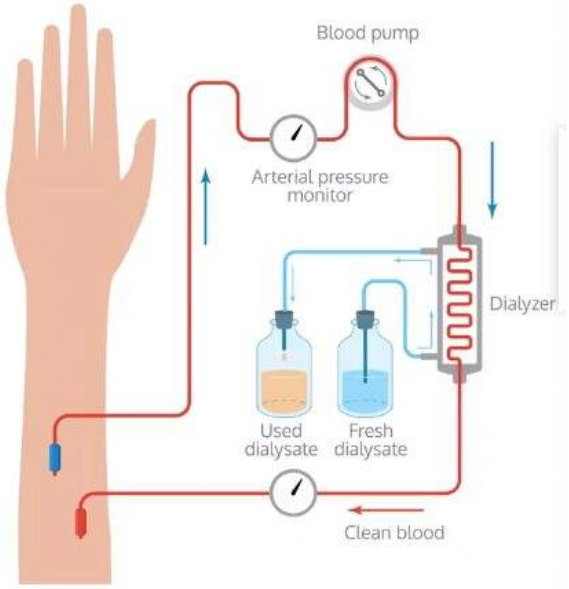
5.2.3 Loss of kidney function: dialysis

Dialysis

- Removes waste from the blood when diseased kidneys are no longer able to effectively clean the blood
- The most common type of dialysis is haemodialysis in which the blood is removed from the body and circulated through a dialyzer

KEY CONCEPTS

- Loss of kidney function can occur for numerous reasons, including diabetes, high blood pressure, kidney infections and blockages.
- Most of these conditions cause damage to the fragile nephrons.
- Dialysis uses an 'artificial kidney' to remove urea from the blood when kidney function is insufficient to do so.
- Dialysis can also balance the water and salt levels to some extent.
- There are two types of dialysis: haemodialysis and peritoneal dialysis.
- Renal dialysis patients undergo dialysis several times a week for extended periods.
- The effectiveness of a technology can be assessed on the basis of certain criteria.



5.3 Effectiveness of technologies

technology	Who is the technology good for
Cochlear implants	Good for individuals that have damaged hair cells in the cochlear
Bone conduction implants	Have a mixed or conductive hearing loss in one or both ears. Are prone to outer and/or middle ear infections. Experienced middle ear trauma
Hearing aids	persons with sensorineural hearing loss (hearing loss in the inner ear due to damaged hair cells or a damaged hearing nerve).

technology	Who is the technology good for
spectacles	People who require vision correction, due to the shape of their lense
Laser eye surgery	improve or correct myopia (short-sightedness), hypermetropia (long-sightedness) and astigmatism (uneven curvature of the eye's surface).

technology	Who is the technology good for
Peritoneal dialysis	Peoples whos kidneys no longer function well, less severe kidney failure
hemodialysis	kidney function drops to 15% or less

5.3.1 effectiveness of laser eye surgery

92-98% of patients are satisfied with the results of lasic surgery
Risks:

- Subconjunctival haemorrhage
- Dry eys

- Follow up surgery
- Glasses or contact lenses
- Chronic physical pain
- Problems with glare during the day or halos, starbusts or other visual aberrations at night
- Flap slippage
- Particles under the flap
- Inflammation at the interface between the flap and underlying stroma
- Post- lasic corneal ectasia- a bulging of the cornea
- Corneal scarring (impossible to wear contacts)

5.3.2 effectiveness of bone conduction implants

- 100% satisfaction rate, bone- anchored hearing aids are extremely effective
- Significantly better hearing for voice to appropriately chosen patients

Verylittle has been published about negative side effects

- Local infection following the operation
- Loss of the osseointergrated fixture as a result of trauma
- Chronic pain
- expensive

5.3.3 effectiveness of cochlear implants

1. Evaluate the use of cochlear implants for a 3 year old child that is profoundly deaf as a result of meningitis.

- A congenitally deaf child should have cochlear implant surgery before 3 years old, earlier if possible.
- Success rate for the cochlear implanted children was 26.87%
- Deafness due to meningitis is a challenging situation because the infection causes sclerosis of the cochlea, which can make cochlear implantation difficult or even impossible. We need to do the implants as early as possible after making the diagnosis before the entire cochlea fills with bone and scar

5.3.4 effectiveness of hearing aids

5.3.5 effectiveness of cataract surgery