Non-Infectious Disease Study Notes

TOPIC 1 - Homeostasis:

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

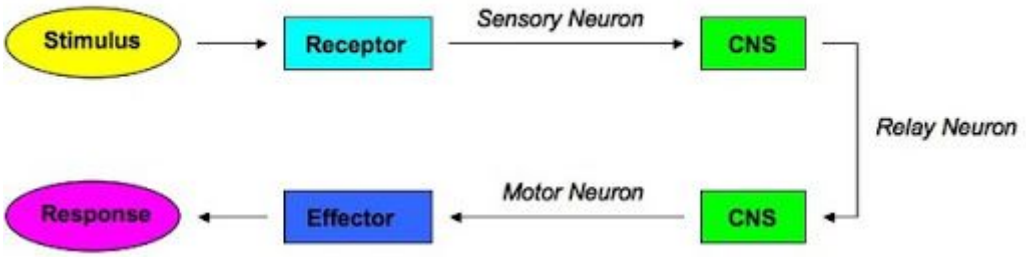
Homeostasis:

- Definition → Homeostasis is the process by which organisms maintain a relatively stable internal environment
- A constant stable environment ensures that cells can survive and generally the chemical processes that occur within the body can only do so within a range of conditions. By having a constant internal environment, the body can function to its fullest capacity and maintain metabolic efficiency.
- Ambient temperature → Temperature of surroundings
- Living organisms need to control:
  - Body temperature
  - Metabolic rate
  - The concentration of dissolved salts
  - The concentration of nutrients (e.g. glucose)
  - Input and output of water
  - Waste products (urea)
  - Oxygen and carbon dioxide levels
- A balance is needed to ensure an organism maintains a stable internal environment to keep their cells balanced. This is essential for many metabolic pathways and the function of the enzymes that control them
- Homeostasis is the process by which organisms maintain a relatively stable internal environment
- Consists of two stages:
  1. Detecting changes from the stable state
  2. Counteracting changes form the stable state

STIMULUS	SENSE ORGANS	SENSES	RECEPTORS
LIGHTWAVES	EYES	VISION, SEEING	RETINA
SOUNDWAVES	EARS	AUDITION, HEARING	COCHLEA
AIR MOLECULES	NOSE	OLFACTORY SMELLING	HAIR CELLS LOCATED IN THE UPPER PART OF THE NOSE
CHEMICAL SUBSTANCES FROM FOOD	TONGUE	GUSTATION TASTING	TASTE BUDS
PAIN, PRESSURE, TEMPERATURE TEXTURE	SKIN	TACTILE TOUCH	OUTERMOSE LAYER-DEAD CELLS MIDDLE LAYER-HAIR FOLLICLES

Stimulus-Response Model:

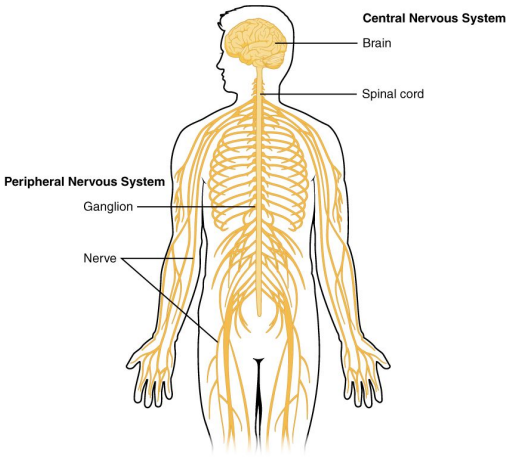
- The human body uses a stimulus-response model to maintain a balance
- Both the endocrine and nervous systems are used for this process
- Steps:
  - A stimulus is detected by the receptor cells (e.g. eyes, skin)
  - A signal is sent to the coordination centre
  - Another signal is sent to the effector cells a
  - A response is invoked



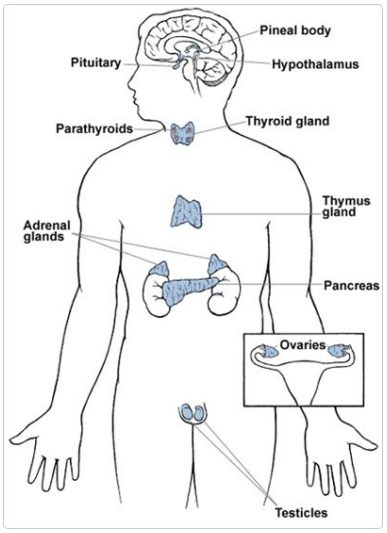
Stage 1 - Detecting a Change:

- The stimulus can be:
  - Internal (levels of carbon dioxide, oxygen, water, wastes and temperature)

    OR
  - External (light, day length, sound, vibration, temperature, texture, odour)
- Receptors are used to detect a change and include the skin, nose, mouth, ears, eyes and muscles
- Types of receptors:
  - Photoreceptors: light
  - Chemoreceptors: detects the concentration of chemicals in the body
  - Thermoreceptors: detect changes in temperature
  - Mechanoreceptors: movement, pressure
  - Osmoreceptors: detect changes in osmotic pressure
- Central nervous system
  - Includes brain and spinal cord:
  - Receives information from the receptors, interprets it and initiates a response by the effectors
  - Sensory, inter-connecting and motor neurons are the cells involved



- Endocrine system:
  - Hormones are chemical messages that are produced by the endocrine glands
  - Hormones travel in the blood and are produced in response to a change in the internal environment of the organism
  - Includes the pituitary gland (releases hormones and regulates the activity of other glands), thyroid gland, adrenal glands and pancreas



Stage 2 - Counteracting the Change:

- Effectors in the body respond to the change to bring the cells back to homeostasis (balance)
- Effectors:
  - Muscles → contract and relax to bring about movement
  - Glands → Secrete chemical substances e.g. Hormones, insulin, aldosterone

1.1 Feedback Loops

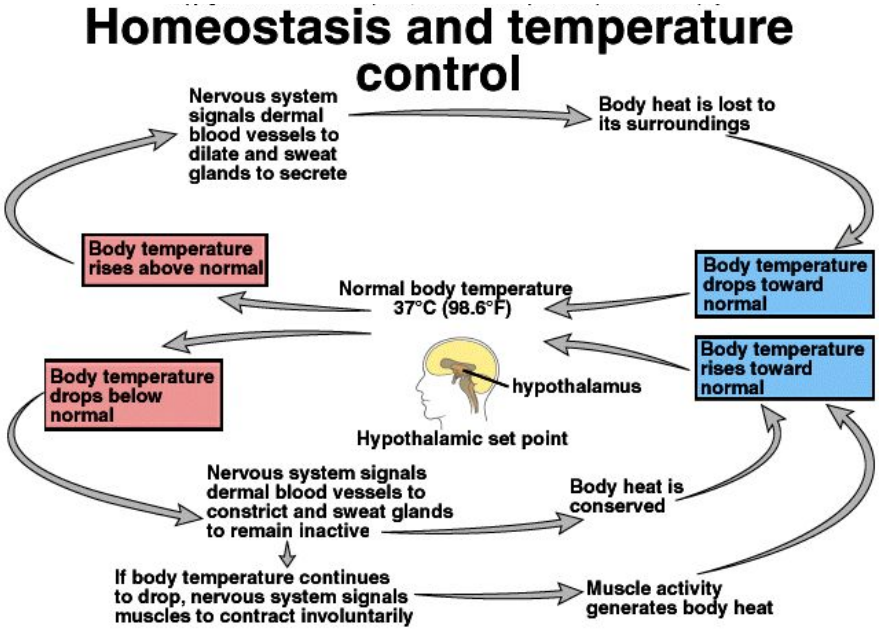
- *Construct and interpret negative feedback loops that show homeostasis by using a range of sources, including:*
  - *Temperature*
  - *Glucose*

Optimal Temperature:

- Most organisms live in environments ranging between 0-45°C depending on the temperature needed for their metabolism
- Mammalian metabolism including enzymes and cell activity is very sensitive to temperature changes
- Optimum internal body temperature for an efficient function is 30-37°C
- Sweating after exercise is an example of something that the body does to bring its temperature back to normal - these adaptations regulate your body temperature
- When your internal temperature changes, sensors in your central nervous system (CNS) send messages to your hypothalamus. In response, it sends signals to various organs and systems in your body. They respond with a variety of mechanisms.
- If your body needs to cool down, these mechanisms include:

- Sweating: Your sweat glands release sweat, which cools your skin as it evaporates. This helps lower your internal temperature.
- Vasodilatation: The blood vessels under your skin get wider. This increases blood flow to your skin where it is cooler — away from your warm inner body. This lets your body release heat through heat radiation.

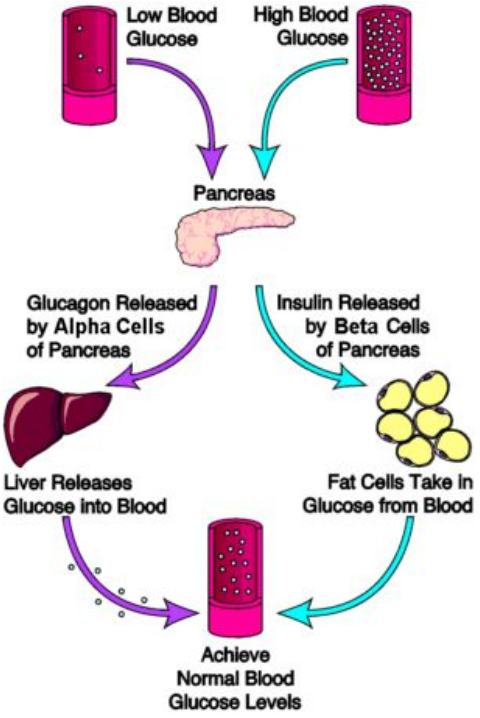
- If your body needs to warm up, these mechanisms include:
  - Vasoconstriction: The blood vessels under your skin become narrower. This decreases blood flow to your skin, retaining heat near the warm inner body
  - Thermogenesis: Your body's muscles, organs, and brain produce heat in a variety of ways. For example, muscles can produce heat by shivering
  - Hormonal thermogenesis: Your thyroid gland releases hormones to increase your metabolism. This increases the energy your body creates and the amount of heat it produces.
- Function:
  - The hypothalamus in the brain monitors your bodies internal temperature (which is blood temperature)
  - If there is a change in stable body temperature then the hypothalamus will initiate responses to increase or decrease your body temperature as needed
  - This is an example of a feedback mechanism

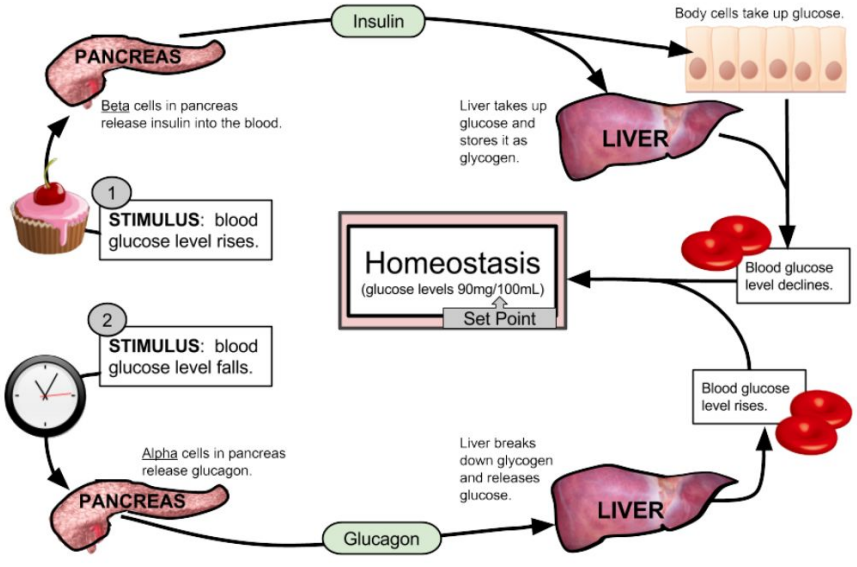


<u>Glucose Control:</u>

- Glucose is the main source of energy for cells so it is important to control the amount being used up and stored
- The pancreas produces insulin and glycogen which regulate the amount of glucose in the blood
- Insulin and glucagon are the hormones which ensure blood sugar (glucose) is maintained.
- It is the production of insulin and glucagon by the pancreas which ultimately determines if a patient has diabetes, hypoglycemia, or some other sugar problem.

- Insulin lowers blood glucose by transporting glucose from the blood into the cells
- Glycogen prevents glucose levels from dropping too low by releasing an enzyme to break down glycogen into glucose
- Insulin is normally secreted by the beta cells of the pancreas. The stimulus for insulin secretion is a HIGH blood glucose. Although there is always a low level of insulin secreted by the pancreas, the amount secreted into the blood increases as the blood glucose rises. Similarly, as blood glucose falls, the amount of insulin secreted by the pancreatic islets goes down.
- Glucagon is secreted by the alpha cells of the pancreatic islets in much the same manner as insulin, except in the opposite direction. If blood glucose is high, then no glucagon is secreted.
- When blood glucose goes LOW, however, (such as between meals, and during exercise) more and more glucagon is secreted. Like insulin, glucagon has an effect on many cells of the body, but most notably the liver.
- The effect of glucagon is to make the liver release the glucose it has stored in its cells into the bloodstream, with the net effect of increasing blood glucose. Glucagon also induces the liver (and some other cells such as muscle) to make glucose out of building blocks obtained from other nutrients found in the body (e.g. protein).





1.2 Homeostatic Mechanisms

- *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*
    - *Internal coordination systems that allow homeostasis to be maintained, including hormones and neural pathways*
    - *Mechanisms in plants that allow water balance to be maintained*

1.2.1 Behavioural, Structural and Physiological Adaptations

<u>Adaptations:</u>

- An adaptation is any characteristic that increases an organism's chance for survival
- There are three types of adaptations for animals:
    1. Behavioural
    2. Structural
    3. Physiological
- An endotherm is an organism that maintains its own body temperature by generating heat from their metabolic processes
- Thermoregulation in endotherms is extremely important because they need to be able to cope with ambient temperature changes while maintaining a stable internal body temperature

<u>Behavioural Adaptations:</u>

- Seeking shade and laying down → For example, Kangaroos will seek out shade and lay on the ground to help them stay cool. This way they are out of the sunlight and the ground is cooler for them to sit on
- Nocturnal behaviour → Daytime is much hotter than night time to many animals are partly or completely nocturnal
- Digging and living in burrows → The bilby and wombat will spend the majority of their day in a burrow. The burrow is underground and much cooler than the surface. Many burrowing animals are also nocturnal
- Evaporative cooling → As our bodies heat up, sweat glands release sweat and it allows the skin to cool down on hot days or when the body temperature increases. Kangaroos lick their forearms which then evaporates in the heat and therefore helps to reduce their body temperature. There is also evaporative cooling in panting. When a dog pants, water is evaporating from the mouth and tongue as the hot air from the planting flows over it
- Huddling together → This conserves body heat and allows them to decrease their surface area to volume ratio and lose less heat to the environment. Penguins commonly huddle together

<u>Structural Adaptations:</u>

- Fur/feathers/hair → This is used to trap air against the skin, causing less airflow at the surface and less heat loss. The thicker the covering the better it is at trapping the air and reducing heat loss in cold climates

- Blubber → A layer of fat under the skin. It is very effective in retaining heat in cold water. Whales can have 60cm of blubber under their skin to help them survive in polar waters.
- Size of ears → Large and thin ears will increase the surface area and allow for more heat loss in hotter climates
- A large SA:V ratio means heat is lost more easily
- A small SA:V ratio means heat is retained easier

Physiological Adaptations:

- Thirst → This indicates that the organism is dehydrated. Water loss due to sweating and evaporative cooling needs to be replaced
- Hibernation → During cooler periods some animals sleep. This allows their body temperature to drop and metabolism to slow down and the animal survives off it's fat reserves. Some animals (e.g. the pygmy possum) can hibernate for up to a year
- Vasodilation → Blood vessels near the surface of the skin become bigger allowing for more heat to be lost to the environment. This is useful in hotter climates
- Vasoconstriction → Blood vessels near the surface of the skin contract and converse heat (e.g. blue lips after swimming)

1.2.2 Internal Coordination Systems

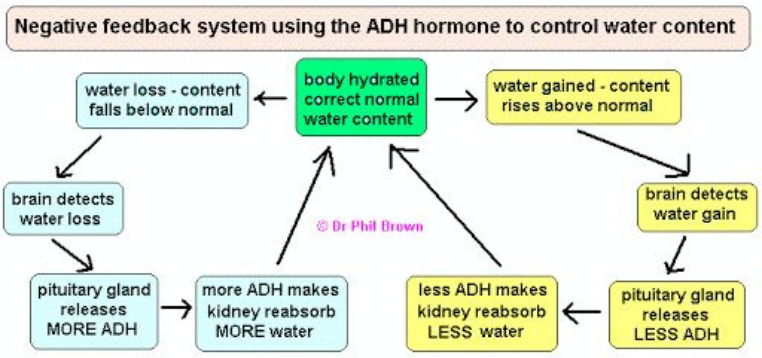
Hormonal Controls:

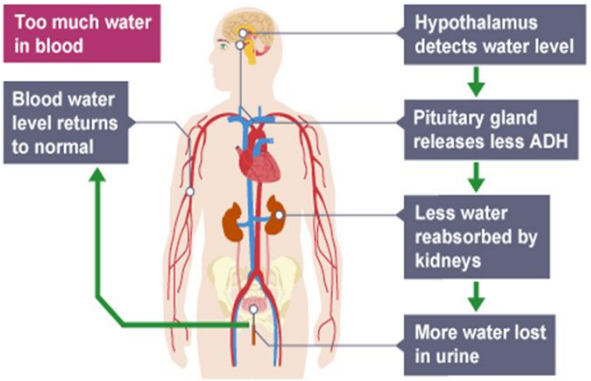
- Hormones are molecules produced by the endocrine glands and travel in the blood to the target cells
- Hormones bind to specific receptors on the target cells and cause change through a process called 'signal transduction'
- Signal Transduction has 3 steps:
  1. Reception → hormone binds to the receptor on the target cell
  2. Transduction → hormone-binding triggers signalling cascade within the cytoplasm
  3. Response → signalling initiates response which is usually transcription of a gene within the nucleus
- Signal Transduction for adrenaline (epinephrine). This shows how binding to surface receptors can change the expression of certain proteins which can then impact on the structure/function of the organism
- Hormones can affect the metabolism of target cells/tissues by increasing or decreasing the activity
- Hormones control many reactions such as digestion, respiration, sleep (melatonin), growth (human growth hormone), reproduction (FSH, LH) and mood

ADH and water control:

- Osmoregulation is the regulation of water and salt in the blood. This is necessary because cells and enzymes work best in an isotonic solution (balanced)
- The hormones Antidiuretic Hormone (ADH) and aldosterone are involved
- Osmoregulation by the kidney → Antidiuretic Hormone controls water reabsorption by the kidneys
- ADH is produced by the hypothalamus and stored in the pituitary gland

- ADH is released into the bloodstream and carried to the kidneys when water levels are too low (e.g. dehydration and after exercise). This increases water reabsorption back into the blood which then produces concentrated urine.
- Negative feedback involved in osmoregulation:



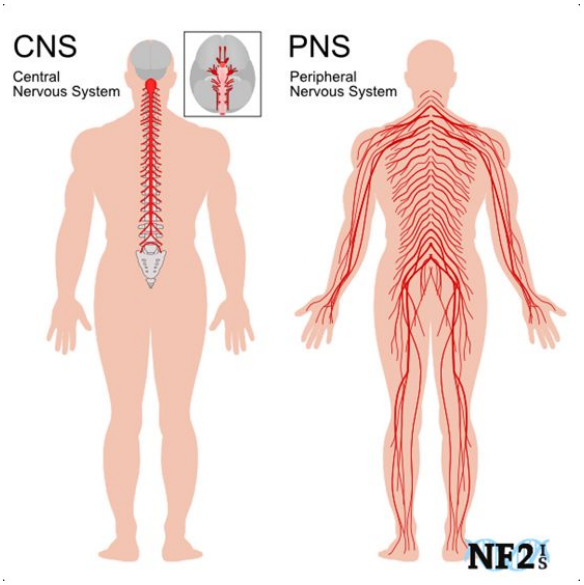


Hormone	Endocrine Gland	Target cell/organ	Effect
Prolactin	Anterior pituitary	Breast gland cells	Milk production
Growth Hormone (GH)	Anterior pituitary	All body cells	GH is vital for normal physical growth in children; its levels rise progressively during childhood and peak during the growth spurt that occurs in puberty.
Antidiuretic hormone (ADH)	Posterior pituitary	Kidney collecting ducts and distal tubule	The antidiuretic hormone helps to control blood pressure by acting on the kidneys and the blood vessels. Its most important role is to conserve the fluid volume of your body by reducing the amount of water passed out in the urine.
Oxytocin	Posterior pituitary	Ducts of breast and uterus	Oxytocin is a hormone that acts on organs in the body and as a chemical messenger in the brain, controlling key aspects of the reproductive system, including childbirth and lactation, and aspects of human behaviour.
Testosterone	Gonads	Testes, body cells	In men, it's thought to regulate sex drive (libido), bone mass, fat distribution, muscle mass and strength, and the production of red blood cells and sperm. A small amount of circulating testosterone is converted to estradiol, a form of estrogen.
Oestrogen	Ovaries	Uterus & body cells	Oestrogen is important for sexual and reproductive development, mainly in

			women.
Thyroxine	Thyroid gland	Body cells	It plays vital roles in digestion, heart and muscle function, brain development and maintenance of bones.
Calcitonin	Thyroid gland	Bone cells and kidneys	Calcitonin is involved in helping to regulate levels of calcium and phosphate in the blood, opposing the action of parathyroid hormone.
Adrenaline and noradrenaline	Adrenal gland	Liver and heart	Prepares the body for fight or flight
Aldosterone	Adrenal glands	Nephron (in the kidney)	Aldosterone affects the body's ability to regulate blood pressure. It sends the signal to organs, like the kidney and colon, that can increase the amount of sodium the body sends into the bloodstream or the amount of potassium released in the urine
Insulin	Beta cells in the pancreas	Body cells	Insulin is released into the bloodstream where it helps to move glucose from the food we have eaten into cells to be used as energy.
Glucagon	Alpha cells in the pancreas	Liver	Stimulate glucose production in the liver and thereby to maintain adequate plasma glucose concentrations

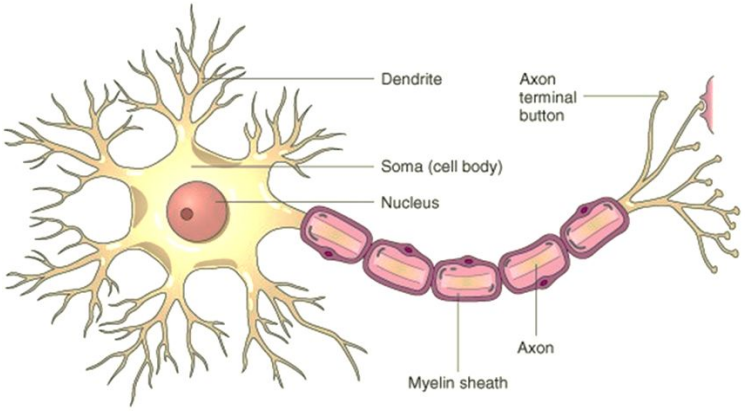
<u>Neural Pathways:</u>

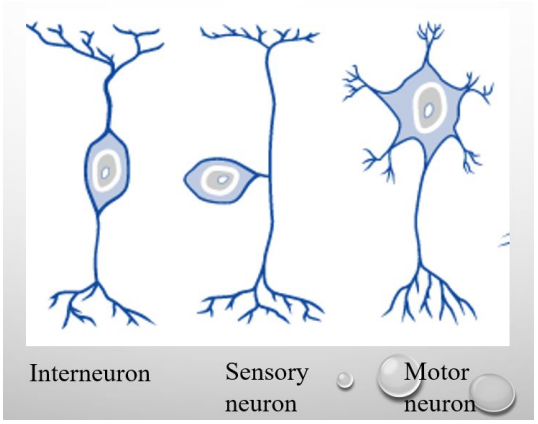
- **Central Nervous System (CNS)** includes the brain and spinal cord → The CNS is a control centre that coordinates responses in the body. It also receives information from and sends responses to the Peripheral Nervous System and correctly interprets and coordinates responses
- **Peripheral Nervous System →** Aids with communication in the body. It passes messages to and from the CNS quickly, allowing the body to respond to the changes
- The nervous system is composed of neurons (nerve cells) which pass electrochemical signals around the body. There are 3 types.
- The movement of this electrical impulse is known as an 'action potential'. When the impulse reaches the end of a neuron it must cross a



synapse to get to the next neuron and it does this by signal transduction and it becomes a chemical message.

- In a neuron, the impulse travels in one direction only. The three types are:
  1. Sensory neurons → transmit impulse from a sense organ to CNS
  2. Interneuron → connects sensory neuron to motor neuron (usually in the brain or spinal cord)
  3. Motor neurons → transmit impulse from CNS to muscle or gland



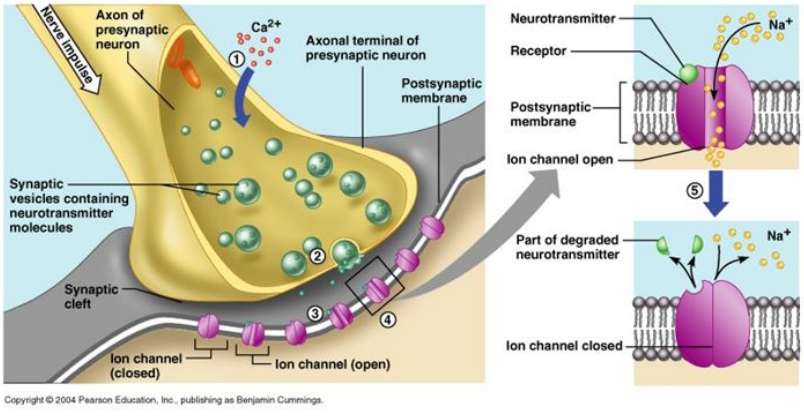


- Neurons are nerve cells that transmit signals by electrochemical changes in their membranes. They consist of a cell body containing a nucleus, cytoplasm and organelles, and extensions or processes to either end called dendrites and axons.
- The neuron transmits an electrochemical signal in one direction only. The dendrites take the signal to the cell body and the axon conducts the signal away from the cell body
- **Axons** → Many neurons only have one axon. Axons branch at the end leading to many synaptic terminals. These release neurotransmitters into the gap between two different neurons. The gap is called the synapse. Some axons have a fatty insulating material known as the myelin sheath wrapped around the axon. The myelin sheath keeps the electrical signal in the neuron so it is transmitted the full length of the neuron.
- **Dendrites** → Small branch-like projections that connect to other cells. The branching nature increases the available surface area and allows the collection of incoming impulses. Dendrites can grow and shrink during the life of the neuron. Alcohol and old age has been shown to reduce dendrites while a situation where the person is learning can increase the growth of dendrites. Dendrites are shorter and thinner than the axon.
- **Synapse →** Between each neuron is a small gap called a synapse. For an impulse to cross the synapse, neurotransmitters are released by the axon terminals. Common neurotransmitters are dopamine, histamine and endorphins. When these chemicals reach the dendrites of the next cell it causes the next neuron to fire. This explains why the signal in neurons is known as an electrochemical response. Along the neuron, the signal is electrical while at the synapse the signal is changed into a chemical signal. The effects of many drugs such as depressants and stimulants affect the transmission of nerve impulses across the synapses of nerve cells by suppressing or increasing the release of neurotransmitters

- **Nerves** → nerves are bundles of nerve fibres (dendrites or axons) outside the brain. The fibres are surrounded by myelin sheath which acts as an insulator. The auditory nerve and optic nerve in the ear and the eye are nerves. These are collective axons coming from the hair cells in the cochlea or the rod and cone cells in the eye.
- Summary of neurons:
  - Axon → the long extension that carries nerve impulses away from the body of the cell.
  - Axon terminals - the hair-like ends of the axon.
  - The cell body → A membrane-bound sections of the neuron that contains the nucleus and other organelles of a neuron.
  - Dendrites → the branching structure of a neuron that receives messages
  - Myelin sheath → the fatty insulating substance that surrounds and protects some nerve fibres
  - Nucleus → the organelle in the cell body that contains the genetic material of the cell
  - Neurotransmitters → these are chemicals that transmit chemicals across a synapse
  - Synapse → gap or space between two neurons

<u>Transmission of a nervous impulse:</u>

- When an atom or group of atoms is electrically charged it is called an ion. Some ions within cells are positive (calcium ion Ca+, sodium ion Na+, potassium ion K+) while others carry a negative charge
- Cells are surrounded by a semipermeable membrane that controls the passage of ions into and out of the cell. In the membrane are ion channels that allow particular ions to pass through
- The difference between the concentration of particular chemical ions inside and outside of a cell results in a negative electrical charge within a neuron
- If a positive charge occurs in a neuron it will be passed to the next neuron as a voltage pulse



- An action potential is a rapid and short change in the membrane potential of an excitable cell. In nerve cells, it causes a nerve impulse while in muscle cells it causes a contraction. Any cells which can have an action potential generated in it is said to be excitable and is able to be made active

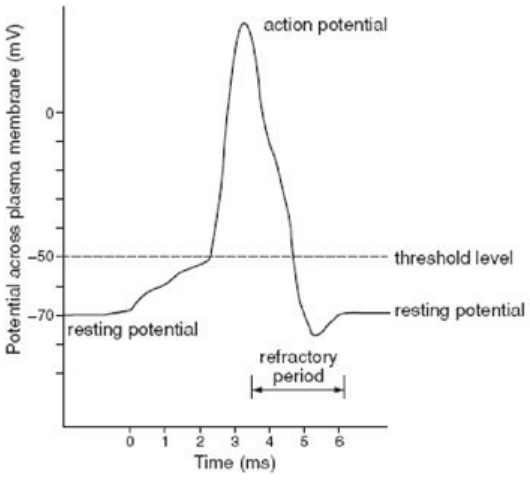
- Different sensory neurons are fired by different stimuli. Different excitable cells have different thresholds for firing. The threshold is the particular value of the membrane voltage that needs to be reached for an action potential to be started

Resting Potential:

- At rest, the inside of a neuron is negative compared to the outside of the neuron
- This charge is obtained by potassium ions (K+) passing easily through the membrane to the outside while chloride ions (Cl-) and sodium ions (Na+) cannot cross freely
- The negatively charged proteins within the neuron are also prevented from crossing the membrane
- As well as ion channels in the membrane there are ion pumps and these use energy to move more ions across the membrane. All of this results in the resting potential being a negative charge of -70 millivolts (mV) within the neuron

Action Potential:

- Refers to how messages travel along the neuron
- An action potential is the name given when a neuron sends an impulse to the next neuron and this occurs when there is a rapid movement of potassium ions and sodium ions across the cell membrane. This is known as depolarization (stage 2 & 3)
- When a neuron is stimulated the first response is for the sodium channels to open and this results in sodium ions rushing into the cell. The positive charge on the sodium ions depolarise the cell and the overall negative charge starts to fall towards zero
- When the negative charge reaches roughly 55 millivolts the neuron will fire and an impulse or spike will occur. This is the threshold for the reaction. It is a non-graded or 'all or none' response
- There is no variation in the response: it either fires completely or not at all. If the stimulus is not large enough to reach the threshold level the neuron will not fire
- Following the opening of the sodium channels, the potassium channels open and potassium moves out of the cell (this is the repolarisation: stage 4) of the cell and results in the cells charge moving back towards the resting potential
- The time taken to return to the resting potential is known as the refractory period and the neuron cannot fire again
- Within the refractory period, there is a stage called the hyperpolarization stage. This occurs when the cells becomes more negative than the normal resting potential (-70mV) as a result of potassium ions (K+) continuing to move out of the cell
- The cell will eventually stabilize at its resting potential and this means it will be ready to carry out more action potential responses to detected stimuli



Summary of steps:

1. The neuron gets stimulated and this stimulation causes a change in the resting membrane potential (-70mV = resting)
2. If a neuron is stimulated enough the inside of the cell will reach a critical level called a threshold (about -55mV)
3. At this point, the sodium ion channels will open (Na +)
4. Sodium ions rush into the neuron because of diffusion forces (high to low) and charge attraction (+ and -). This causes the voltage to rise. This is depolarization
5. The charge inside the cells eventually reaches about +30mV. This is relative to the outside of the cell, the inside is now positive and the outside is negative. At this point, the sodium ion channels close. This change in polarization is called depolarization
6. Next, potassium ion channels open up. This causes K+ to rush out of the cell
7. As the K+ leaves it causes the inside of the cell to become negative again (-70mV) and this is referred to as repolarisation

1.2.3 Balancing Water Levels in Plants

- Plants may be adapted in ways which allow them to balance water levels
- Mechanisms for water retention are observed widely in Australian flora as they are often subject to harsh, drought-like conditions
- Theory of transpiration-cohesion-tension theory: the mechanism by which water flows through the xylem of plants is due to the combined effects of:
  - Transpiration: evaporation of water through the stomata of plants
  - Cohesion: that water molecules are attracted to each other so will move in a cohesive stream
  - Tension: water molecules are attracted to the surfaces when they touch
- By regulating levels at which transpiration occurs, plants are able to retain or release water as required. This can happen through:
  - Smaller leaves
  - Closing stomata
  - Movement
  - Dropping leaves during summer or drought
  - Large cavities for water storage

<u>Homeostasis in Plants:</u>

- The stomata → These are the pores on a leaf and regulate the exchange of gas and water
- Guard cells → The cells surrounding the stomata that regulate whether they are opened or closed
- Plants live in a diverse range of environments and so they have different adaptations to help them cope
- There are three groups of plants:
  1. Hydrophytes
  2. Mesophytes
  3. Xerophytes

Hydrophytes:

- These live in freshwater
- There is a large amount of water available to them but it also means that due to osmosis, water is constantly moving into the plant tissue and diluting the internal tissue concentrations
- Many of these plants have a high number of stomata on their leaves to help with removing the excess water and maintaining the internal balance
- Example → Water lily

Mesophytes:

- Land plants with adequate and regular water supply
- Medium-density of stomata on leaves that assist in photosynthesis and water balance in the plant
- These plants are not drought tolerant as they are accustomed to regular water supply
- Example → corn, palm trees

Xerophytes:

- Land plants that live in dry climates and have a diverse range of adaptations to help them conserve water
- Their leaves have a small surface area
- Usually have a thick waxy cuticle
- Their leaves hang vertically to reduce the surface area that is exposed to the sun
- Extensive root system to obtain water from a wider area
- Storing water in stems (e.g. like cacti and succulent plants)
- Reduced leaves into spine and stems are photosynthetic

TOPIC 2 - Causes and Effects:

Inquiry Question: Do non-infectious diseases cause more deaths than infectious diseases?

2.1 Overview of Causes and Effects

- *Investigate the causes and effects of non-infectious diseases in humans, including:*
  - *Genetic diseases*
  - *Diseases caused by environmental exposure*
  - *Nutritional diseases*
  - *Cancer*

2.1.1 Genetic Diseases

Cause:

- Gene or chromosomal abnormality caused by mutations
- Genetic disorders can be caused by a mutation in one gene (monogenic disorder), by mutations in multiple genes (multifactorial inheritance disorder), by a combination of gene mutations and environmental factors, or by damage to chromosomes (changes in the number or structure of entire chromosomes, the structures that carry genes).

Effect:

- To function correctly, each cell depends on thousands of proteins to do their jobs in the right places at the right times. Sometimes, gene mutations prevent one or more of these proteins from working properly. By changing a gene's instructions for making a protein, a mutation can cause the protein to malfunction or to be missing entirely. When a mutation alters a protein that plays a critical role in the body, it can disrupt normal development or cause a medical condition. A condition caused by mutations in one or more genes is called a genetic disorder.
- In some cases, gene mutations are so severe that they prevent an embryo from surviving until birth. These changes occur in genes that are essential for development, and often disrupt the development of an embryo in its earliest stages. Because these mutations have very serious effects, they are incompatible with life.
- It is important to note that genes themselves do not cause disease—genetic disorders are caused by mutations that make a gene function improperly.

Example - Down Syndrome:

- Down's syndrome is a genetic disorder caused when abnormal cell division results in extra genetic material from chromosome 21.
- Down's syndrome causes a distinct facial appearance, intellectual disability and developmental delays. It may be associated with thyroid or heart disease.
- Other symptoms include:
  - Developmentally → delayed development, learning disability, short stature, or speech delay in a child
  - Eyes → lazy eye or spots
  - Also common → difficulty thinking and understanding, brachycephaly, upslanting palpebral fissures, atlantoaxial instability, bent little finger, congenital heart disease, displacement of the tongue, excess skin on the back of the neck, flaccid muscles, hearing loss, immune deficiency, low-set ears, mouth breathing, obesity, obstructive sleep apnea, polycythemia, seborrheic dermatitis, single line on palm, thickening of the skin of the palms and soles, thyroid disease, or vision disorder

Incidence:

- The best estimate available is that there are between 13,000-15,000 individuals with Down syndrome in Australia as of 2019. This estimate is based on Western Australia data and applying these numbers to the Australian population.
- These population figures mean that for every 10,000 people there are 5.14 people with Down syndrome.
- It is estimated that approximately 1 in every 1100 babies born in Australia will have Down syndrome. This means that each year there are approximately 290 new babies born each year who have Down syndrome.
- The incidence of births of children with Down syndrome increases with the age of the mother. The chance of a woman conceiving a child with Down syndrome varies from 1 in 1400 for a woman 20 years of age to 1 in 30 at age 45 years. Younger women have babies more frequently, so the majority of babies born with Down syndrome are born to women under 35 years of age.

Prevalence (in Australia):

- In Australia DS is identified in 1:1150 live births (around 260 births each year).
- The chance of a woman conceiving a child with DS varies from 1 in 1400 for a woman 20 years of age to 1 in 32 at age 45 years.
- The chance of having a child with DS increases with maternal age,
- The prevalence of people with DS in our community has increased over recent decades due to the increase in life expectancy and changes to care practises.

Mortality:

- Sometimes infants with Down syndrome are born weighing less than 1,500 grams, or about 3.3 pounds. This is called "very low birth weight." These babies are 24 times more likely to die in the first 28 days of life compared to infants with Down syndrome of normal birth weight (between 2,500 grams and 4,000 grams, or between 5.5 and 8.8 pounds).
- Infants with Down syndrome who also had a congenital heart defect (CHD) were five times more likely to die in the first year of life compared to infants with Down syndrome who did not have a CHD
- The number of infants with Down syndrome that die before one year of age has declined over time. For example, between 1979 and 2003, among babies with Down syndrome, the rate of death during the first year of life declined from 8.5% to 5.0%, a decrease of about 41%. For comparison, the rate of death during the first year of life among all babies in the general population declined from 1.5% during 1979-1983 to 0.9% during 1999-2003, a decrease of about 40%.

Treatment and Management:

- Primary care providers to monitor growth, development, medical concerns and provide vaccinations.
- Speech therapists to help them communicate.
- Physical therapists to help strengthen their muscles and improve motor skills.
- Occupational therapists to help refine their motor skills and make daily tasks easier.
- Behavioural therapists to help manage emotional challenges that can come with Down syndrome.
- Surgery for heart defects and other physical disabilities that may require attention

2.1.2 Environmental Exposure Diseases

<u>Cause:</u>

- Environmental diseases (ENVDs) are non-communicable diseases that result when people are chronically exposed to toxic environmental chemicals. Other contributory causes of ENVDs include radiation, pathogens, allergens and psychological stress.
- Various causes include:
  - Lifestyle diseases are caused by people's choices, such as smoking, alcohol, food choice, and exercise (or lack of it). These include heat disease, lung cancer, type 2 diabetes, and obesity.

- Chemical exposure can be due to lack of knowledge about the chemical's danger, such as with asbestos and mesothelioma, or it can be accidental such as lead poisoning.
- Allergens are elements which only cause disease/immune response in certain individuals, and this can range from mild hay-fever to anaphylaxis. Allergens include food, plant materials, and the most common allergen, dust.
- Other elements of the physical environment can also cause disease – for example UV radiation causing skin cancer etc.

Effect:

- There can be various effects on a person
- Certain environmental caused disease can lead to cancer and often death

Example - Melanoma:

- Melanoma occurs when the pigment-producing cells that give colour to the skin become cancerous.
- Caused by excessive exposure to ultraviolet (UV) light from the sun, solariums or radiation from nuclear substances
- The UV light causes changes to the DNA of the skin cells which may cause continued abnormal cell division. This can lead to the formation of malignant melanomas
- It is the most serious type of skin cancer
- Symptoms include: bigger mole diameter, darkening of the skin, mole color changes, or skin mole with an irregular border

2.1.3 Nutritional Diseases

Cause:

- Nutritional diseases are caused by diets lacking the proper balance and amount of nutrients
- Can also be caused by psychological conditions that lead to inappropriate diets
- This imbalance in the diet leads to a condition known as malnutrition
- There are two categories of malnutrition:
  1. Undernutrition - insufficient intake of the correct type of food, a quantity of food or a combination of both
  2. Overnutrition - excessive intake of food

Effect:

- The major causes of death, illness and disability in which diet and nutrition play an important role include coronary heart disease, stroke, hypertension, atherosclerosis, obesity, some forms of cancer, Type 2 diabetes, osteoporosis, dental caries, gallbladder disease, dementia and nutritional anemia.

Example - Type 2 Diabetes:

- Type 2 diabetes is a disorder where the body becomes resistant to insulin and gradually incapable of producing it. This means glucose builds up in the bloodstream which can damage many-body systems. Both genetic and environmental factors are involved.

- Causes include lack of exercise, unhealthy food consumption, family history
- Symptoms:
    - Being excessively thirsty
    - Passing more urine
    - Feeling tired and lethargic
    - Always feeling hungry
    - Poor wound healing
    - Itching, skin infections
    - Blurred vision
    - Gradually putting on weight

Prevention methods include:

- Management of weight
- Exercise regularly
- Eating a balanced, healthy diet
- Limiting takeaway and processed foods
- Limiting alcohol intake
- Quitting smoking
- Controlling the blood pressure.

Treatment and Management:

- Insulin injections to help lower blood glucose
- If the patient is obese, weight loss is recommended to help reset metabolism - this may require extreme measures such as gastric band surgery
- Possibly, diabetes medication or insulin therapy.
- Monitoring blood sugar levels

Future Research:

- Prevention measures - improving public understanding through high impact education programs
- Studies to look at genetic risks and identifying genes that put people at risk of developing type 2 diabetes
- Studies into the production of insulin - to make it cheaper and more accessible to the low-socioeconomic members of society who are often more affected due to diet/lifestyle
- Research into pancreas transplantation - where a healthy pancreas is transplanted and could produce insulin
- Islet cell transfer (instead of the whole pancreas) may be able to restore function in the pancreas and would be less invasive than a whole organ transplant

2.1.4 Cancer

Cause:

- Uncontrolled cell growth due to many different factors including exposure to mutagens, genetic factors, mutagens and some viruses
- Cancer is caused by accumulated damage to genes. Such changes may be due to chance or to exposure to a cancer-causing substance. The substances that cause cancer are called carcinogens. A carcinogen may be a chemical substance, such as certain molecules in tobacco smoke.
- Symptoms: some patients have abnormal bumps, unexplained fevers, night sweats or unintentional weight loss.

Effect:

- Cancer happens when cells that are not normal grow and spread very fast. Normal body cells grow and divide and know to stop growing.
- Over time, cells die and a growing tumour becomes a lump of cancer cells that can destroy the normal cells around the tumour and damage the body's healthy tissues.
- Most cancer survivors experience some side effects as a result of their diagnosis or treatment. The type of side effects depends on your diagnosis, treatment type, and overall health. Some of the most common side effects for cancer survivors include fatigue, loss of appetite, nausea, pain, and weight loss.

Example - Breast Cancer:

- Cancer that forms in the cells of the breasts.
- Breast cancer can occur in women and rarely in men.
- Symptoms of breast cancer include a lump in the breast, bloody discharge from the nipple and changes in the shape or texture of the nipple or breast.
- There are various treatments that can be used (depends on the stage of cancer):
    - Surgery (surgical removal of lymph node, surgical removal of breast tissue, plastic surgery to change the size of breast)
    - Chemotherapy (unwanted reactions to drugs given for the purpose of killing cancer cells)
    - Teletherapy (radiation therapy that uses x-rays or other high energy beams to destroy cancer cells and shrink tumours)

Incidence:

- Germline mutations in the BRCA1 gene are associated with elevated risks of breast and ovarian cancer (1,2).
- The absolute risk of cancer by the age of 70 years conferred by a BRCA1 mutation is reported to be between 45% and 87% for breast cancer and between 36% and 66% for ovarian cancer

Prevalence:

- BRCA1 mutations are seen in 7% of families with multiple breast cancers and 40% of families with breast and ovarian cancer.
- Women with a BRCA1 mutation have a 40% lifetime risk of developing ovarian cancer.

Mortality:

- For BRCA1 mutation carriers, we observed 52 deaths (20 from BC) in the surveillance group, and 10 deaths (one from BC) after BRRM.
- The hazard ratios were 0.40 (95% CI 0.20–0.90) for overall mortality and 0.06 (95% CI 0.01–0.46) for BC-specific mortality.

Future Research:

- Ongoing studies are looking at ways to enhance current breast cancer screening options. Technological advances in imaging are creating new opportunities for improvements in both screening and early detection. One new technology is 3-D mammography, also called breast tomosynthesis. This procedure takes images from different angles around the breast and builds them into a 3-D-like image. Although this technology is increasingly available in the clinic, it isn't known whether it is better than standard 2-D mammography, for detecting cancer at a less advanced stage.
- It is now known that breast cancer can be divided into subtypes that respond differently to various types of treatment. The three main clinical subtypes of breast cancer are:
  - Hormone receptor (HR) positive. HR-positive breast cancers are those that contain the estrogen receptor (ER) and/or progesterone receptor (PR). These cancers grow in response to these hormones and can be treated with hormone therapies.
  - Human epidermal growth factor receptor 2 (HER2) positive. HER2-positive breast cancers are those that have high amounts of the HER2 protein; they can be HR positive or HR negative. These cancers can be treated with therapies that target HER2.

2.2 Incidence, Prevalence, Mortality

- *Collect and represent data to show the incidence, prevalence and mortality rates of non-infectious diseases, for example:*
  - *Nutritional diseases*
  - *Diseases caused by environmental exposure*

<u>Definitions:</u>

Incidence → The rate of new cases of the disease. It is generally reported as the number of new cases occurring within a period of time and is usually reported as a fraction of the population at risk of developing the disease (e.g. per 100,00)

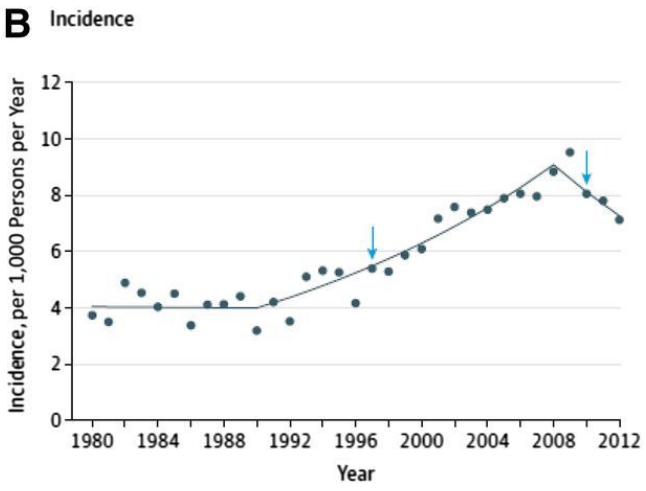
Prevalence → The actual number of cases alive with the disease either during a period of time or at a particular date in time. Period prevalence provides a better measure of the disease load since it includes all new cases and all deaths between two dates, whereas point prevalence only counts those alive on a particular date.

Mortality → A measure of the number of deaths from the disease in a particular population, scaled to the size of that population, per unit of time

2.2.1 Nutritional Disease - Type 2 Diabetes

Incidence:

- Around 17,400 people with type 2 diabetes began insulin treatment in 2017, equating to around 4,100 cases per 100,000 population.
- According to the NDR: Incidence rates for insulin-treated type 2 diabetes were 1.5 times as high in females as males (5,100 compared with 3,300 per 100,000, respectively)



Prevalence:

- The prevalence of diabetes has tripled between 1989–90 and 2014–15.
- The proportion of people with diabetes has increased from 1.5% to 4.7%.
- In 2014–15, the prevalence of type 2 diabetes among adults:
  - Was higher for men (7%) than women (5%).
  - Increased rapidly up to age 75, with rates among 65–74 year-olds (17%) 3 times as high as for 45–54 year-olds (5%) and 1.4 times as high as for 55–64-year-olds

Mortality:

- In 2016, Type 2 diabetes was the cause of 4,770 deaths, making it the 7th leading cause of death overall
- The death rate from diabetes was 16.2 deaths per 100,000 people.

2.2.2 Environmental Disease - Melanoma

Incidence:

- In Australia, the age-standardised incidence rate for melanoma increased by 93% between 1982 and 2015, from 27 cases per 100,000 persons to 52 cases per 100,000 persons (and was estimated to remain at this level in 2019)
- Melanoma incidence increased between 1982 and 2002 but after this point rates of melanoma diagnosis somewhat stabilized.
- Melanoma is much more common in males and possible reasons for this are: they are more likely to work outdoors thus exposed to more UV radiation and they are also less likely to use suncream or other protection against UV.

Prevalence:

- Prevalence is the actual number of people who are still living with the disease during a given time frame. These numbers are commonly used to calculate the 5-year survival rate which is shown in this graph.
- The 5-year survival rate hasn't improved much since 1987 but women have a higher survival rate, possibly because they are more likely to undergo regular skin checks and therefore receive an earlier diagnosis.

Mortality:

- Mortality rates are much higher in men than they are in women. This makes sense because more men are diagnosed with melanoma and therefore they are more likely to die from the disease.
- From around 2012, there is a relatively steep decline for the mortality of melanoma. This is probably due to two factors:
    1. First, new treatment such as immunotherapy which has been shown to be effective in around 50% of cases means fewer people are dying from melanoma.
    2. Second, the declining mortality rate suggests that public health programs have been effective in creating awareness and have encouraged people to get skin checks and treatment sooner rather than later.

TOPIC 3 - Epidemiology:

Inquiry Question: Why are epidemiological studies used?

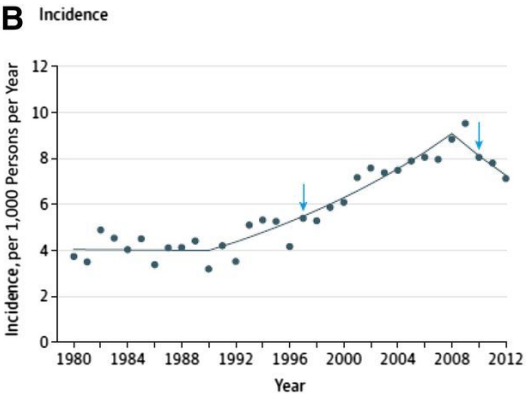
3.1 Patterns of Non-Infectious Disease

- *Analyse patterns of non-infectious diseases in populations, including their incidence and prevalence, including:*
    - *Nutritional diseases*
    - *Diseases caused by environmental exposure*

3.1.1 Nutritional Disease - Type 2 Diabetes

Incidence:

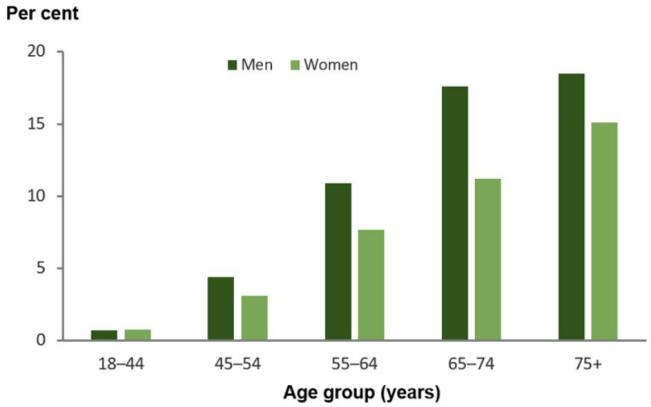
- Around 17,400 people with type 2 diabetes began insulin treatment in 2017, equating to around 4,100 cases per 100,000 population.
- According to the NDR: Incidence rates for insulin-treated type 2 diabetes were 1.5 times as high in females as males (5,100 compared with 3,300 per 100,000, respectively)
- Due to the increase in globalisation and fast/cheap food being more accessible it is easier for people to develop poor diet habits and become overweight. This is in comparison to previously when people may have been more likely to spend more time doing outdoor activities and cooking/eating fresh food (i.e. more complex carbohydrates and less fat)



Prevalence:

- The prevalence of diabetes has tripled between 1989–90 and 2014–15.
- The proportion of people with diabetes has increased from 1.5% to 4.7%.
- In 2014–15, the prevalence of type 2 diabetes among adults:
  - Was higher for men (7%) than women (5%).
  - Increased rapidly up to age 75, with rates among 65–74 year-olds (17%) 3 times as high as for 45–54 year-olds (5%) and 1.4 times as high as for 55–64 year olds
- Diabetes prevalence has been rising more rapidly in low- and middle-income countries than in high-income countries.
- Older adults are at high risk for the development of type 2 diabetes due to the combined effects of increasing insulin resistance and impaired pancreatic islet function with aging.

Figure 1: Prevalence of self-reported type 2 diabetes, among persons aged 18 and over, by age and sex, 2017–18



Analysis:

- Nutritional transition is a term used to define a set of changing risk factors that a country may face as they develop
- Changes in diet: as countries become more developed, populations usually begin to purchase more processed food rather than grow or buy fresh ingredients meaning diets have more fat and lower complex carbohydrates
- Changes to lifestyle: as food production technologies develop, populations undergo changes to their work and leisure activities usually resulting in more sedentary lifestyles

3.1.2 Environmental Disease - Melanoma

Incidence:

- In Australia, the age-standardised incidence rate for melanoma increased by 93% between 1982 and 2015, from 27 cases per 100,000 persons to 52 cases per 100,000 persons,(and was estimated to remain at this level in 2019)
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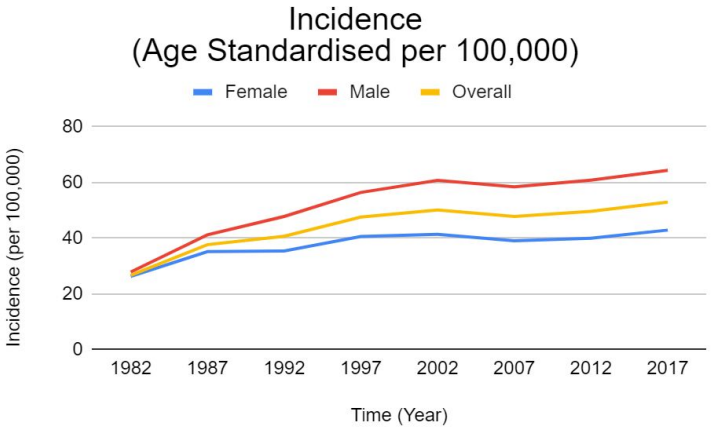

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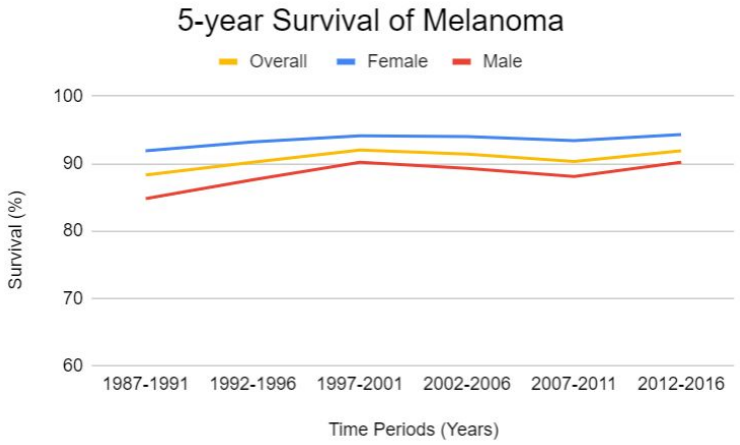


3.2 Treatment, Management and Research

- *Investigate the treatment/management, and possible future directions for further research, of a non-infectious disease, using an example from either a nutritional disease or a disease caused by environment exposure*

3.2.1 Environmental Disease - Melanoma

Prevention:

- Spending limited time in the sun to avoid UV radiation
- Avoiding sunburn

- Avoid using solariums
- Protect skin/body when exposed to UV radiation (e.g. hat, sunscreen)
- Getting moles/skin regularly checked
- Making sure the population is informed about the risks associated with the disease

Treatment:

- The treatment options for melanoma are surgery, chemotherapy and radiation therapy as well as newer methods which are personalised for the patient.
- These are targeted therapy and immunotherapy which can eliminate tumours entirely in some patients.
- The treatment used depends on the severity and duration of the melanoma, that is, how long a person has been suffering from the disease and whether it has metastasised to other parts of the body. When melanoma is detected earlier, the treatment is more likely to be effective.
- Types of treatment/description:
  - Surgery → To either remove the melanoma or to remove affected lymph nodes.
  - Chemotherapy → This method uses drugs to kill cancer cells. Chemotherapy can be given intravenously or in a pill form.
  - Radiation therapy → Uses high-powered energy beams to kill cancer cells. Radiation therapy may be directed to the lymph nodes if the melanoma has spread to that area. Radiation therapy can also be used to treat melanomas that can't be removed completely with surgery.
  - Targeted therapy → Focus on specific weaknesses present within cancer cells. By targeting these weaknesses (and using targeted drugs) cancer cells die. Usually recommended when cancer has spread to lymph nodes or other areas in the body
  - Immunotherapy → A drug treatment that assists the immune system in fighting cancer and is commonly used after surgery when melanoma has become more severe. Immunotherapy treatments can sometimes be injected directly into the melanoma.

Future Research:

- Melanoma peptide vaccines are being evaluated in clinical trials for patients with both localized and advanced melanoma.
- Research has shown that vaccination can cause the immune system to fight melanoma, even in advanced disease, but these therapies are still considered experimental.
- Targeted therapy and immunotherapy are undergoing trials and there is continual research into ensuring this goes smoothly
- Other types of immunotherapy are being researched, for example, adoptive cell therapy (ACT). ACT involves identifying those immune cells from a patient that are best at recognizing their tumour, growing a lot of those cells in the lab, and giving them back to the patient.
- There are continual public health programs and government research in ensuring people are made aware of the risk and potential disease contraction

3.3 Methods and Benefits of Epidemiology

- *Evaluate the method used in an example of an epidemiological study*
- *Evaluate, using examples, the benefits of engaging in an epidemiological study*

3.3.1 Methods of Epidemiology

Epidemiology:

- Definition → the study of the occurrence, prevalence and spread of disease within a population and to use the study to control or manage health problems
- Epidemiological studies can look at both infectious and non-infectious diseases (can also be car accidents, suicides etc)
- Epidemiological studies are used to demonstrate a relationship between a disease and environmental factors revealing possible risk factors and causative agents
- These studies can then provide information and strategies to deal with the spread or containment of disease in a population
- Epidemiology involves the collection and analysis of very large quantities of data

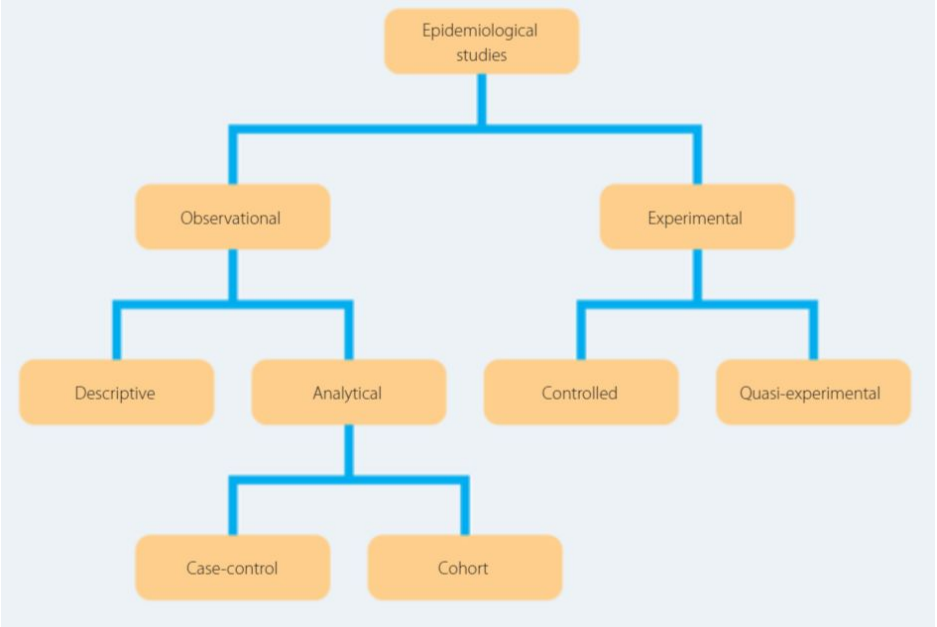
Features of Epidemiology:

- Large quantities of data analysed
- Information is gathered from different sources
- Information related to the disease is studied across an extended period of time
- Information about other factors in the environment is compared with information about the disease
- Participants should represent the wider population
- Control groups (who are not exposed to the potential cause of disease but are similar to the test group should be used
- Data on incidence, prevalence, mortality and morbidity should be used
- A possible cause and risk factors should be identified
- The effectiveness of control and treatment programs should be studied

Types of Data:

- Age
- Gender
- Education and job status
- Geographical location
- Incidence → number of new cases
- Prevalence → number of people affected at any one time
- Morbidity → number of cases as a proportion of the population
- Mortality → the number of deaths

Types of Studies:



Descriptive → shows patterns of distribution of diseases in occupations e.g. geographical distribution, occupational groups, socioeconomic groups. It is usually the first investigation that is conducted and it includes:

- A hypothesis
- Who has the disease?
- Where is the disease found?
- In what groups is the disease found?
- When is the disease found (e.g. age, developmental stage)
- How many people does the disease affect?

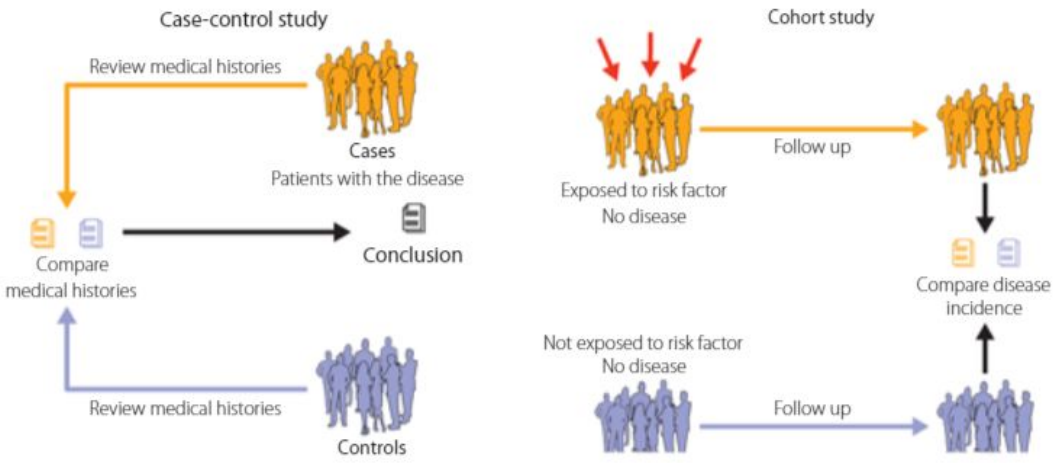
- Example: In determining the cause of lung cancer, the data collected included information about the age, sex, smoking habits, diet, occupation and drinking habits of people with and without lung cancer

Analytical → investigations to test hypotheses e.g. comparison of the lifestyle of people with a disease to identify common risk factors (case-control studies and cohort studies are types of analytical studies). It includes:

- Risk factors can be identified (through establishing correlation or association between risk factors and the incidence of the disease)
- Potential causes can be identified (through establishing correlation or association between risk factors and the incidence of the disease)
- Cause and effect is established
- Morbidity
- Mortality
- Incidence
- Prevalence

- Case-Control studies compare people with the disease (case) to people without the disease (control) and then look for differences in possible exposure to the disease

- Cohort studies involve studying two or more similar groups of people who no longer have the disease. The groups differ in one main factor: their potential exposure to the disease



Intervention → measures the effectiveness and safety of interventions e.g. clinical trial of a new drug. The aim of an intervention treatment is to change the behaviour of the population as a whole in order to reduce the incidence of the disease. It includes:

- The effectiveness of preventive and control measures that have been implemented to prevent and control the disease respectively
- An example is an experimental study which is often used to test the effectiveness of a new type of drug. In this study, people who are suffering from a particular condition are observed for a set time period. Participants are placed into two groups where one receives a drug and the other receives a placebo. The effects of the drugs are records and statistically analysed to determine the effectiveness of the drug being studied. Meaning that a vaccine for influenza could be tested in hospital workers. One department would be given the vaccine and the others not (the incidence of influenza would then be compared between the two groups)

<u>Errors:</u>

- Sometimes there are errors in epidemiological studies. They can either be random errors or systematic errors
- Random Errors:
  - Unpredictable variations in the data that impact the results of the study
  - They can make the study less precise but they don't create bias. The best way to avoid this is a large sample size and studying separate groups
- Systematic Errors (bias):
  - Selection bias is when there is bias in selecting participants for the study causing the data to deviate or lean from the true value e.g. sampling bias: when a population or group are over or under-represented in the study (such as gender of age)
  - Information bias involves errors made when measurements or recording information occurs e.g. measurement bias: measurements are inconsistently or

inaccurately recorded. This is why measurements are repeated and protocols are followed

3.3.2 Evaluation of Epidemiological Studies

Evaluation:

- To evaluate involves making a judgement about something and using evidence to support the judgement. The evidence should be based on specific criteria relevant to the particular scenario being evaluated
- The validity of the method used in an epidemiological study should be evaluated on whether it follows accepted epidemiological principles for the study that is being carried out
- Criteria common to most epidemiological studies include large sample size, long period of study, the use of scientifically approved methods of conducting the study, collecting data and analysing results
- Other criteria will depend on the type of study (e.g. whether it is a case-control, cohort or experimental study)

Benefits:

- The primary benefit which arises from epidemiological studies is to discover or provide substantial evidence for significant risk factors associated with a non-infectious disease
- Identify who has the disease, where the disease is found, in what groups and when is the disease found (e.g. age, developmental stage)
- It will also be possible to identify the risk factors and potential causes of the disease. This is because the study allows us to establish a correlation or association between risk factors and the incidence of the disease. This allows us to analyse the potential cause and effects of the disease
- Intervention epidemiology can determine the effectiveness of preventive and control measures that has been implemented to prevent and control the disease respectively
- Identifying areas of strength and weaknesses for a population's health. For example, Australia has a high life expectancy for both males and females but also has an increasing number of people getting diagnosed with type 2 diabetes
- Allows identification for the major causes of mortality and morbidity across Australia and smaller population groups such as people of low socioeconomic status or Aboriginal and Torres Strait Islanders
- It gives us information about how people use our healthcare system and help outbreaks in certain diseases or illnesses
- Vital for your understanding of health priorities in Australia
- It is used to guide what is focused on for health promotion, who the health promotion targets, where the health promotion is needed, how the health promotion is delivered and where to deliver it
- Measuring the degree to which different risk factors are present in the crisis affected community and the degree to which populations benefit from interventions to minimize these risk factors

- Monitoring trends in the occurrence of specific diseases over time and identifying emergent threats in a timely fashion. For example, studies can be conducted in specific geographical areas where a number of people become affected with certain diseases
- Discovering the agent, host and environmental factor that affect community health to provide scientific basis for prevention of disease and injury including health promotion.
- Identifying at risk population at greater risk of specific diseases
- To evaluate effectiveness of health programmes and services

<u>Example - The Pima Indian population:</u>

- Various studies have linked increased physical activity with a change in body mass and composition. These factors contribute to a reduced incidence of type 2 diabetes
- A cohort study was carried out on the Pima Indian population (a native American population living in Arizona)
- This study examined the role of physical activity in the development of type two diabetes. This study was carried out between 1987 and 2000 with 1728 non-diabetic Pima individuals aged between 15 and 59
- Method:
    - The participants were interviewed about their physical activity. The interview was conducted by trained personnel who implemented a valid and reliable questionnaire
    - Each participant's leisure and occupational activity for the past year was assessed sepearley. Only physical activity that was greater than normal everyday tasks were used
    - From a list of common acitibites the participants were asked to record their leisure activities (over the past year) including duration and frequency
    - To measure occupational activity the participants were asked to report on all the jobs they had had in the past year. For each job the average work schedule, time spent walking/riding to work, the number of hours spent sitting and the most common activities that were performed when not sitting were recorded
    - Activities were then weighted according to their intensity (using scientifically accepted models). Estimates were calculated separately for both the leisure and occupational activities as hours per week and averaged over the previous year. The activity level of each individual was classed as either high or low
    - The interview would judge if a participant was incapable of reporting their activity correctly and if this was the case, their results were excluded from the analysis
    - Incidence rates were calculated by age group, gender and physical activity using a scientifically approved model
    - The results of this study indicated that in most age groups and for males and females the incidence rate of diabetes was lower in those who had higher levels of physical activity than in those who had low levels of physical activity
- Evaluation:
    - The study size of 1728 individuals and the 13-year time period over which it occurred satisfy the epidemiological requirements of a large sample size and a long period of study therefore reducing the effect of sampling bias

- In a cohort study, two or more similar groups of people who are free of the disease should be studied. The major difference between the groups should be the factor that is being studied. This study does this as non-diabetic Pima Indians from the same designated areas and aged 15-59 were studied. The groups varied in the amount of physical activity that was apart of their daily life
- The diagnosis of diabetes was made by objective means → scientifically approved testing at the first and each follow-up visit. This reduced the likelihood of measurement tbias
- Trained interviewers used a scientifically valid questionnaire to determine the activity levels of participants. The trained interviewers and objective questionnaire reduced both interviewer and measurement bias
- Using mathematical models, participants' physical activities were weighted for their intensity level and the activity levels of each individual were then classified as either high or low
- Scientifically tested models were used to analyse the results and determine the incidence rates of diabetes related to activity levels and BMI index
- Data was excluded from analysis if individuals were thought to have incorrectly reported activity levels which reduced recall bias
- The written report of the study was peer reviewed before publication

- Benefits:
  - Gives data about type 2 diabetes
  - Allows organisations or government to implement programs or further studies if there is found to be an issue with diabetes
  - The risk and potential cause of the disease can also be identified and further research can be done
  - Data is now accessible to the wider population and can be analysed in further studies

<u>Example - Skin Cancer in a Subtropical Australian Population:</u>

- The study was done to test the hypothesis that the prevalence of skin cancers (specifically SCC and BCC) in outdoor workers is greater than that in indoor workers.
- Nambour was chosen because previous studies had shown that Nambour has a very high prevalence of BCC and SCC skin cancers (one of the highest anywhere).
- Various special surveys have been undertaken in Nambour, a typical subtropical community in Queensland, Australia.
- Estimates of incidence reported here are based on skin cancers medically treated between 1985 and 1992 and new cases diagnosed by dermatologists in two examination clinics in 1986 and 1992.
- Although as expected, fair skin, a history of repeated sunburns, and nonmalignant solar skin damage diagnosed by dermatologists were strongly associated with both types of skin cancer, outdoor occupation was not.
- Method:
  - In 1986, a random sample of 3,000 individuals were chosen from the 5,100 persons aged 20-69 years listed on the electoral roll as residents of Nambour, Queensland, and were invited to participate in a skin cancer survey

    - A total of 2,095 adults attended an initial skin cancer survey in December 1986, an overall 70 percent response from the electoral sample of 3,000 and a 78 percent response rate among permanent residents
    - Experienced dermatologists examined all participants for prevalent skin cancer on the head, neck, and upper limbs; and a random sample (10 percent) received whole-body skin examinations. Of these residents, 1,770 participated in a November 1987 postal survey regarding any skin cancer treatments in the 2-year period of December 1985 to November 1987
    - The observations recorded by the dermatologist also included subjects' hair color, number of raised and flat nevi on the hands and arms, and signs of solar skin damage (number of solar keratoses on each anatomic site, telangiectasia of face, nuchal elastosis)
    - A standardized questionnaire was used to ask subjects about skin color and present and past occupations (whether mainly outdoors, indoors, or a mixture of indoors and outdoors)
    - Each participant was also asked about typical outdoor recreational exposure, usual number of hours spent outdoors, number of painful sunburns experienced in their lifetime, level of education, parents' countries of origin, smoking and recreational exercise habits, and dietary intake
    - Incidence was used throughout the study, although prevalence was recorded in the 1986 study, as part of a baseline for the study.
- Evaluation:
    - By following up a random sample of nonresponders, the survey population was shown to be representative of the Nambour population with respect to distribution of major risk factors for skin cancer
    - An experienced dermatologist was used to provide an accurate assessment of the skin cancer on those being studied
    - The flaws in selecting people randomly from the electoral roll include that people who choose to participate, are more likely to be those who: are unemployed or retired or have time to spare, are socially oriented, and/or are concerned about skin cancer. There was an attempt to compensate for these flaws by following up with a random sample of non-responders.
    - The control for the study was those who worked indoors
    - All skin cancers reported by study participants were verified against histologic or clinical records, and only confirmed reports were included in the calculation of incidence rates
    - Follow-up time and skin cancer incidence were accumulated for all individuals who responded in a reporting period and were distributed according to their contributions to 10-year age-specific bands. Calculated incidence rates were based on newly affected persons, discounting any follow-up time beyond the incidence of the first lesion in the study period (i.e., subsequent skin cancers were not counted). Incidence rates were age adjusted using the world population standard
- Results:
    - There was no statistically significant correlation between outdoor occupation and the incidence of BCC and SCC skin cancer.

- It also showed that there was a correlation between BCC and SCC cancer and complexion, freckling on the back, and cutaneous sun damage (presence of solar keratoses).
- The lack of evidence for correlation between occupation and skin cancer may be due to a lack of causation
- It may have been due to a systematic tendency for fair-skinned people to avoid outdoor work (This is known as a selection bias).
- It may have been because outdoor workers are more likely to wear hats and sunscreen all the time.

TOPIC 4 - Prevention:

Inquiry Question: How can non-infectious diseases be prevented?

4.1 Disease Prevention Methods

- *Use secondary sources to evaluate the effectiveness of current disease-prevention methods and develop strategies for the prevention of a non-infectious disease, including:*
    - *Educational programs and campaigns*
    - *Genetic engineering*

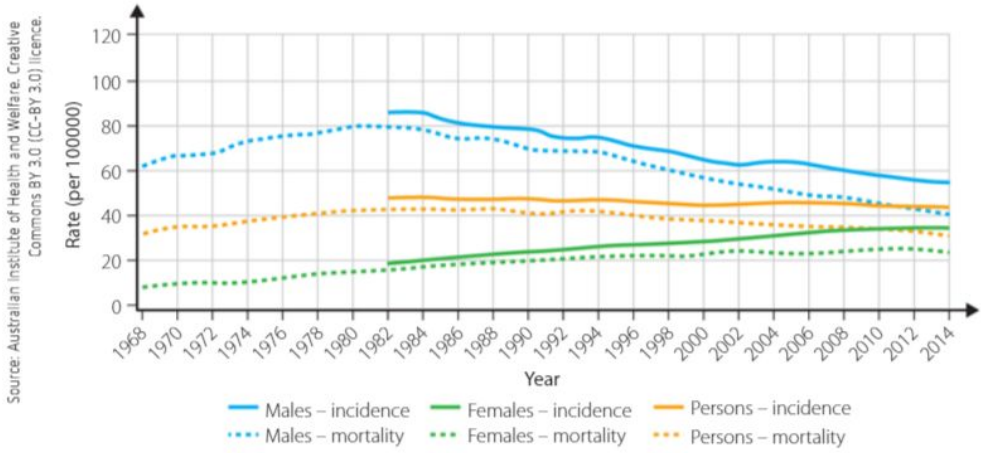
4.1.1 Educational Programs and Campaigns

- Educational program → program that educates the public about the effects of the disease, risk factors and strategies to prevent the disease
- Campaign → a range of strategies used to try and reduce the incidence of the disease
- Through advocating the risks there is a potential to change behaviours and therefore lower the incidence of the disease
- Non-infectious diseases are a major cause of death, disease and disability in the population. This places strain on the health system and economic develop and also affects the wellbeing of many individuals
- The main risk factors associated with non-infectious disease are tobacco, alcohol, unhealthy diet and lack of physical activity
- Educational programs and public health campaigns are often not enough to change the behaviour of a population therefore it is essential that governments introduce legislation to minimise the risk factors in the population
- Various components of public health campaigns include:
    - National Days or weeks to raise awareness and educate people about a disease
    - Online resources to assist individuals and groups in implementing lifestyle behaviour changes
    - Advertising campaigns that saturate all forms of media
    - National helplines i.e. Quitline
    - Support programs such as dietary and physical activity advice
    - Legislation to reduce risk factors
    - Funding for organisations

Lung Cancer Program - QUIT:

- Some of the legislation that has been introduced by the government to reduce the use and effects of tobacco include:
  - A ban on all tobacco advertising, promotion and sponsorship
  - A ban on smoking in the workplace and in public places
  - Plain paper packaging of cigarettes, containing pictorial, graphic warnings and no logos, colours, brand images or promotional information
  - An increase in taxes on tobacco products
- Smoking remains the leading preventable cause of death in the Australian population, with 15,500 smoking-related deaths each year
- The QUIT campaign was developed as an educational program and campaign to reduce the prevalence of smoking related diseases in the population
- Lung cancer is just one of many smoking related diseases which include many types of cancers, heart disease and other lung diseases
- There are studies which show the clear link between cigarette smoking and reduce life expectancy
- Studies comparing smokers and non-smokers show that smokers are 10 times more likely to die from lung cancer
- Further studies show that the more cigarettes smoked each day, the greater the incidence of lung cancer
- The campaign to reduce smoking and smoking related diseases has a multifaceted approach that involves education to raise awareness of risk factors and legislative changes
- The QUIT program has evolved over many years and runs in conjunction with many strategies including:
  - The use of slogans such as ‘Quit for life’ and ‘iCanQuit’
  - Graphic images in the media and on cigarette packets to highlight the dangers of cigarette smoking
  - A national helpline to support smokers in their goal to changing behaviour and breaking the habit
  - Legislation requiring plain paper packaging on all cigarette packets and storage of cigarette packers in cupboards out of sight of consumers
  - Increase excise on tobacco products to make them more expensive
  - Legislation to restrict advertising
- The graph below shows changes in the prevalence of smoking in the Australian population from 1990 to 2013 in relation to the strategies implemented to control tobacco products
  - The graph indicates that strategies implemented to control the prevalence of smoking in the Australian population have been somewhat successful as the rate of smoking reduce from just over 24% in 1991 to 13% in 2013
  - It is predicate that the prevalence of smoking will decrease further with continuation of these strategies
  - Therefore the effectiveness of the campaigns can be demonstrated as there is a clear link between the campaigns and the lowering incidence
  - The combination of both educational programs and campaigns show a decrease in smoking, therefore, displaying its success

- Incidence and mortality rates for lung cancer:
  - The graph below shows an initial increase in the mortality rate from lung cancer for males from 1968 to 1982, followed by a decrease in the mortality rate from 1982 to 2014
  - It also shows a gradual increase in the mortality rate from lung cancer for females from 1968 until the rate becomes steady in 2002
  - There has been a decrease in the incidence rates for males include 1982
  - The female incidence rate of lung cancer was much lower initially but showed a steady increase until 2014
  - The difference in trend between males and females is possible because lung cancer takes decades to develop meaning this can be explained by the decrease in the rate of smoking which occurred earlier in males than in females



4.1.2 Genetic Engineering

- Many non-infectious are the result of mutations in the genetic material
- If scientists are able to trace the causes of disease to specific changes in our DNA sequences, this opens up the possibility of curing diseases using genetic engineering
- Genetic engineering techniques allow us to make changes to the genetic code thus allowing us to edit sequences causing non-infectious disease
- A number of genetic engineering techniques may allow for the prevention of non-infectious disease

<u>Gene Therapy:</u>

- The correction of genetic disorders by introducing a normal. Functioning gene into cells
- This is achieved by inserting correct gene sequences into a cell where a defect has occurred
- Techniques used to inject new genetic material include the insertion of viral vectors, use of a gene gun and inorganic nanoparticles
- Gene therapy can be used to treat:
  - Haemophilia
  - Parkinson's disease

CRISPR-Cas9:

- A gene-editing system by which point mutations may be accurately introduced into genomes
- CRISPR-Cas9 may be used to improve gene therapies by making point mutations to dysfunctional cells
- Such changes will alter the genomes of the edited cells as well as any cells which form from them, resulting in a lasting somatic cell
- The process includes two main elements, a Cas9 enzyme and guide RNA
    - The Cas9 enzyme contains 'molecular scissors' that allow it to cut DNA. the location of DNA that Cas9 cuts is determined by the sequence of the guide RNA
    - This guide RNA contains a pre-designed sequence that will bind to a corresponding DNA sequence, guiding Cas9 to the desire location to cut in the DNA
    - After Cas9 has cut a section of DAn, the cell will detect DNA damage and try to repair it
    - DNA repair is an error-prone process so it has an increased probability of a mutation occurring

Embryo Screening:

- This process increased access and decreased costs of whole-genome sequencing and opens up the possibility of screening embryos for genetic disorders
- This may be particularly beneficial for couples who carry known genes for disease
- Screening allows election of embryos only without the genetic defects for implantation (i.e. this happens through IVF)
- Editing technologies, especially at the early embryo stage, open up the possibility that whole organism changes may be made to the offspring
- IVF can screen for chromosomal abnormalities. It can also screen for certain specific genetic diseases e.g. cystic fibrosis.
- The technologies involved in preimplantation screening include: A few cells from each embryo are taken before implantation and tested. This testing can involve chromosome testing using Next Generation Sequencing to check for chromosomal abnormalities. Karyomapping can also be used to screen for some specific single-gene disorders.

TOPIC 5 - Technologies and Disorders:

Inquiry Question: How can technologies be used to assist people who experience disorders?

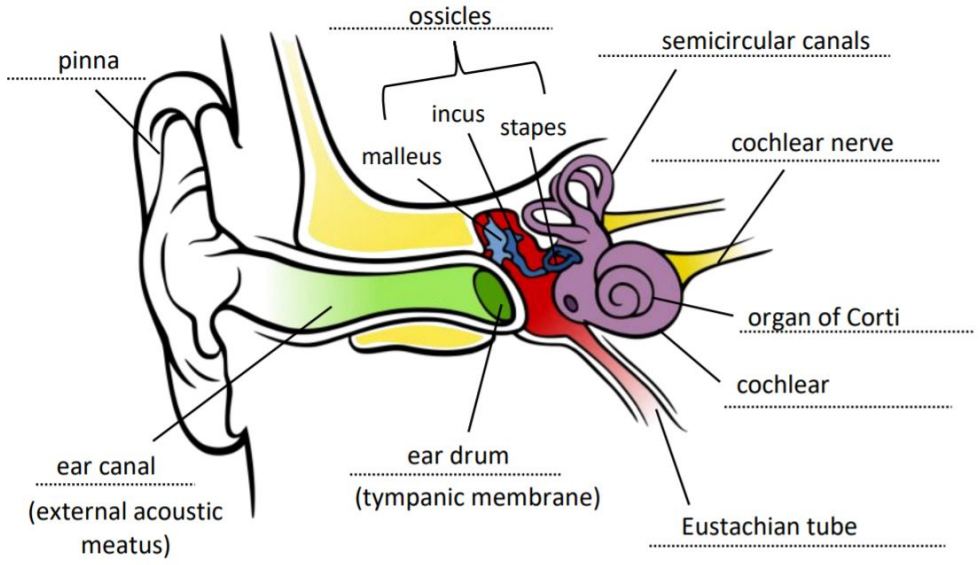
5.1 Cause of Disorders

- *Explain a range of causes of disorders by investigating the structures and functions of the relevant organs, for example:*
    - *Hearing loss*
    - *Visual disorders*
    - *Loss of kidney function*

5.1.1 Hearing Loss

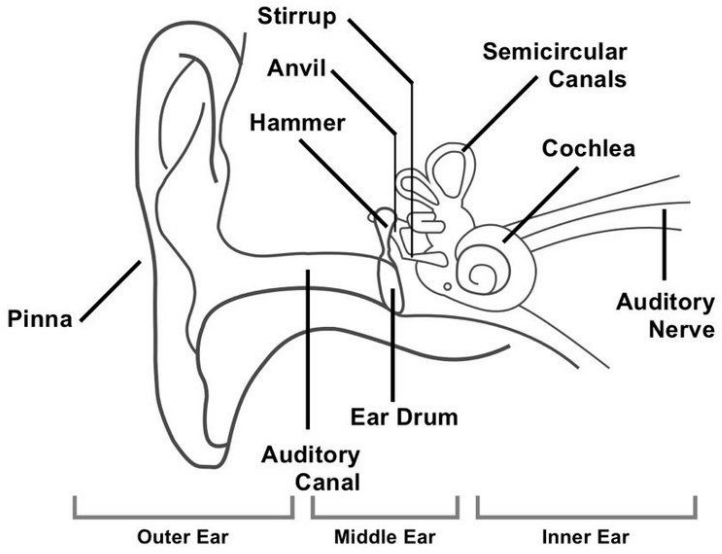
<u>The Ear:</u>

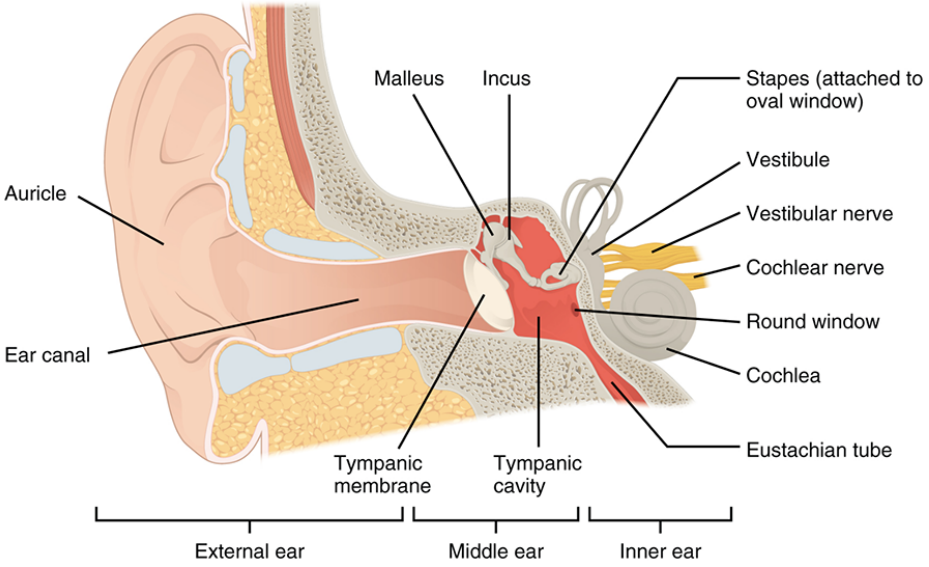
- Our ears are supposed to detect sound and maintain physical balance
- There are three regions of the ear:
    - The outer ear
    - The middle ear
    - The inner ear



- Outer ear:
    - Pinna: fleshy outer part of the ear
    - Auditory canal: leads to the eardrum (tympanic membrane)
    - The eardrum separates the outer ear from the middle ear
    - The eardrum is a thin flap of skin that is stretched tight like a drum and vibrates when sound hits it. These vibrations move the tiny bones of the middle ear, which send vibrations to the inner ear.
- Middle ear:
    - Made of the 3 smallest bones in the body (this is called the Ossicles)
    - Malleus (hammer)

- Incus (anvil)
- Stapes (stirrup)
- The hammer is connected to the eardrum
- Vibration in the eardrum sends a signal through the ossicles
- The stirrup is attached to the oval window and this vibrates, pushing the fluid in the cochlea to move





- Inner ear:
  - The cochlea is a snail-shaped structure filled with fluid
  - The function of the cochlea is to transform the vibrations of the cochlear liquids and associated structures into a neural signal.
  - Semicircular canals and vestibule
  - The organ of Corta contains the receptor cells (hair cells) for sound
  - Audacity nerve contains sensory neurons that take the message to the brain

Hearing:

1. Sound is generated as a longitudinal wave (e.g. ringing a bell)
2. Vibrating air is collected by the pinna and travels down the ear canal
3. Eardrum starts vibrating at the same frequency. Energy is transferred to ossicles.
4. Ossicles vibrate causing fluid in the inner ear to vibrate and then the membrane vibrates
5. Fluid presses on membranes causing a force to be exerted on hair receptor cells in the organ of Corti. Hair cells in the organ of Corti are pulled on
6. Hairs release neurotransmitters into space between neurons leading to auditory nerves transferring impulse to the brain
7. Interpreted as sound

Eustachian Tube:

- This connects the middle ear to the pharynx (throat)
- It equalises pressure inside the ear to the outside environment (e.g. on planes and diving)

The organ of Corti:

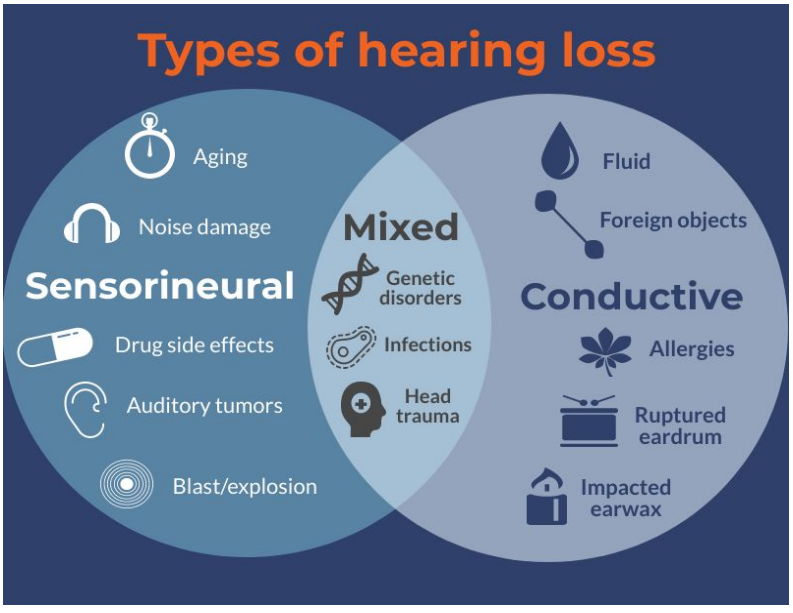
- Sits on top of the basilar membrane in the middle chamber of the cochlea
- It is made up of 15,500 hair receptor cells
- If these hair cells die, they are not replaced. This is why damage to your hearing is irreversible
- Young people will have more hair cells in the organ of Corti because with age, naturally, we lose them
- Hearing also depends on the amount of damage done from loud sound (i.e. concerts and music)
- Sounds with different frequencies have a different pitch
- Pitch is determined by certain hair cells in different regions stimulated within the organ of Corti
- Hair cells at the base have short fibres that vibrate at high frequency
- Hair cells at the apex have long fibres and are stimulated at low frequencies

Damage to hearing:

- Sound is measured in decibels (db)
- 0db = fainest audible sound to humans
- 60db = conversation
- 130db = physically painful (e.g. jet plane, loud concerts)

Hearing Impairment:

- There are two main types of hearing loss:
  - Conductive hearing loss
  - Sensorineuarl hearing loss



Conductive:

- Caused by a problemwith the mechanical conduction of vibrations through the outer and middle ear
- This can occur due to :
  - Malformation of structures in the oter and middle ear
  - Hardenining of the stapes bone due to extra bone
  - Ear infections
  - Wax accumulation in ear canal
- All these factors inhibit the movement of the vibration in the outer and middle ear
- This means that is this volume of the sound that is effected in this type issue
- A hearing aid will magnify the sound vibrations enabling better transmissioninto the ear
- Bone conducting implants will bypass the damaged or malformed parst of the ear and transfer the vibrations directly into the inner ear

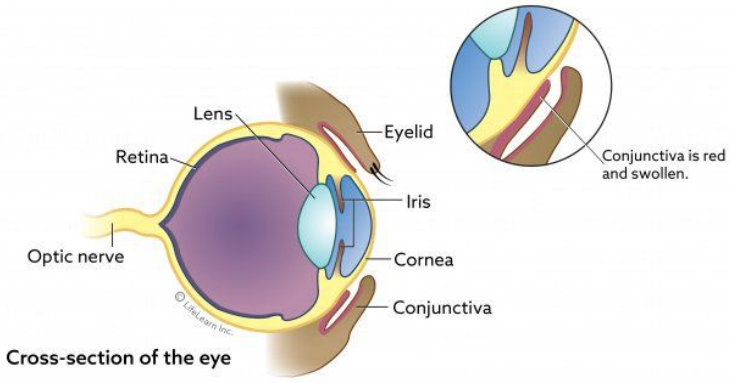
Sensorineaural:

- Cuased by damageg or malformation of the inner ear cincluding the cohclea, hair cells (organ of corti) or the auditory nere
- This can be caused by:
  - Excessive noise exposure
  - Birth defect
  - Infection or tumoour
  - Heredity
- Hearing aids can be used in some situations
- Cochlear implants are used for most inner ear damage where no hearing is left

5.1.2 Visual Disorders

The Eye:

- The eye is responsible for vision and balance
- Light comes into the eye, it is refracted (bent) and focused onto receptor cells in the retina
- The receptor cells send a message on the brain and an image is interpreted
- Humans have two eyes for binocular vision
- The two eyes move in a coordinate way to perceive stimuli (light)
- The eyeball has 3 layers beneath a thin lining called the conjunctiva



Cross-section of the eye

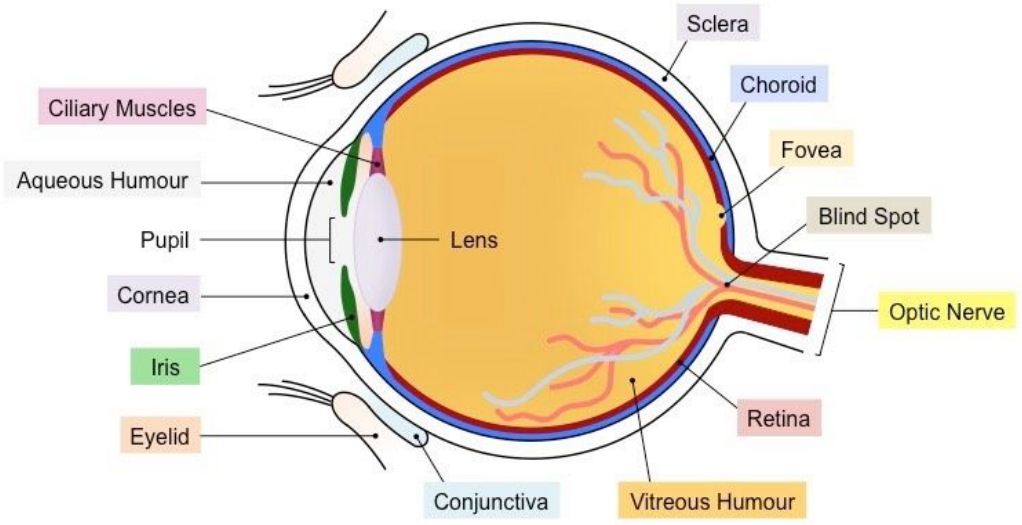
Parts of the Eye:

Structure	Description	Function
conjunctiva	layer of clear epithelial cells across front surface of eye and continuing inside upper and lower eyelids	protects front of eye
cornea	transparent membrane over front of eyeball, made from collagen protein, covers iris and white of eye	protects eye, blocks rays shorter than 300 nm wavelength, refracts light rays to direct them through the pupil into the lens
sclera	white of the eye, continuous with the clear cornea, the tough layer encompasses the whole eyeball	protects eye, holds fluid and keeps eye in a spherical shape
choroid	black layer between sclera and retina with many blood vessels; at the front of the eye the choroid becomes the iris	provides blood supply for the retina, black pigment prevents false images by absorbing stray light, forms the iris
aqueous humour	clear thick liquid held between the cornea and lens	lubricates cornea and lens, helps hold eyeball shape, refracts light
iris	extension of choroid, coloured muscular ring around pupil	muscles contract or dilate to regulate size of pupil, colour pigment blocks excess light
pupil	opening at centre front of eye, circular in humans (can be other shapes, e.g. slit in some species)	changes size to control amount of light entering lens
lens	biconvex flexible disc made of cells with clear crystallin protein	blocks UV rays, changes shape to focus light rays onto retina

- Conjunctiva:
  - A transparent, mucous membrane that shields the sclera
  - Protects the internal structures of the eye and helps lubricate & nourishes the eye with mucus
- The outer layer - Sclera:
  - The sclera is the outermost layer of the eye, the opaque white part at the posterior
  - Made of non-elastic, tough fibrous tissue
  - Function to preserve shape and protect inner layers of the eye
- The outer layer - Cornea:
  - The cornea is a transparent structure at the anterior part of the eye
  - The curve in the cornea helps light to bend or converge so they land in the back of the eyeball
- The middle layer - Choroid:
  - Located in the posterior choroid
  - Dark layer inside the sclera which contains many blood vessels
  - The function is to absorb the prevent light from scattering
- The middle layer - Anterior Choroid:
  - Connects choroid with lens and contains ciliary muscles and suspensory ligaments that hold the lens in position
  - The function is to alter the shape of the lens
  - The forward section of the choroid layer forms the ciliary body, lens and iris
  - The ciliary body is a circular structure in the eye, consisting of ciliary muscles and suspensory ligaments
- Lens:
  - The lens is a transparent protein disc and focuses light into the retina
  - Iris is a pigmented muscular tissue that controls the size of the pupil and therefore the amount of light entering the eye
  - The pupil is an opening in the iris that allows light to enter the eye
- Inner layer - Retina:

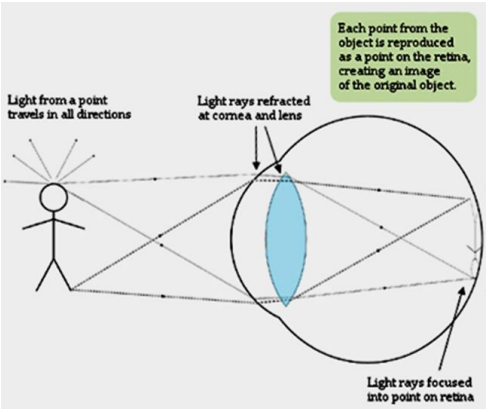
- An inner coat that contains the photoreceptor cells (rods and cones), blind-spot and fovea (where the sharpest image is found)
- There are more rods than cones and they are highly sensitive to shades of black and white and work well in low light
- Cones are colour receptor cells (malfunction = colour-blindness) and give us acuity (sharpest image possible) and work well in full light
- The function is to receive light stimulus
- The retina can be viewed using an ophthalmoscope

- Vitreous and aqueous humour:
  - Aqueous humour is a clear, runny fluid between the cornea and the lens of the eye
  - The vitreous humour is a jelly-like, clear fluid between the lens and the retina that refracts light and helps to maintain the shape of the eyeball
- Optic nerve:
  - The optic nerve is located in the back of the eye. It is also called the second cranial nerve or cranial nerve II.
  - It is the second of several pairs of cranial nerves. The job of the optic nerve is to transfer visual information from the retina to the vision centres of the brain via electrical impulses.



<u>Distortion due to refraction of light:</u>

- When light moves from one medium to another e.g. water to air it is bent or refracted meaning it slows down in a more dense medium
- This refraction of light leads to distortion of that image making it difficult to judge the exact location of the object
- Light needs to enter the eye in parallel lines to make a clear and accurate image

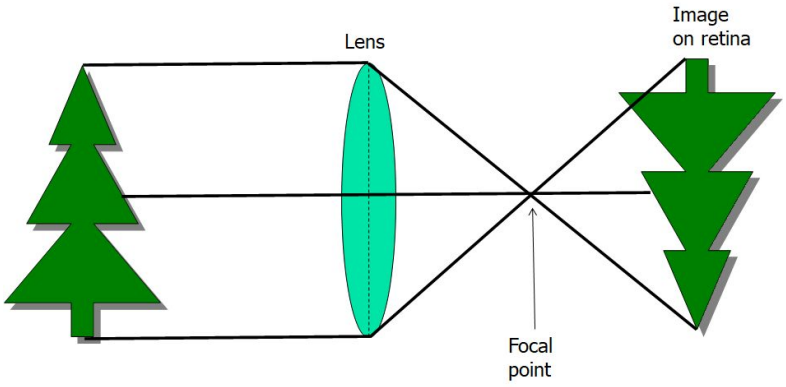


<u>Light in the eye:</u>

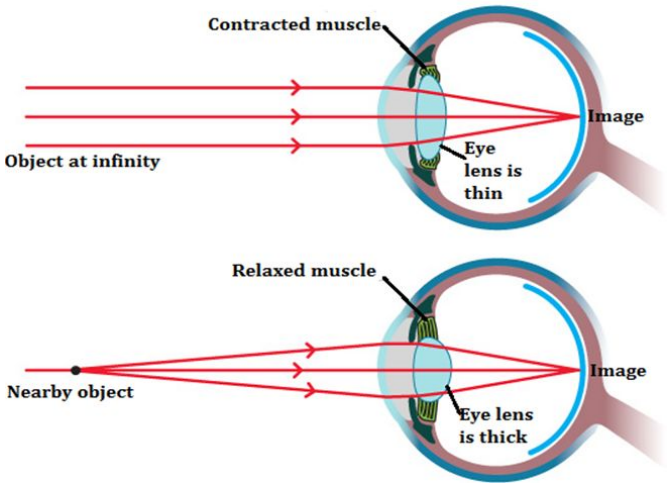
- As light enters the eye it is refracted
- The cornea and lens have the biggest impact and converge the light rays onto the focal point in the retina

Accommodation:

- The changing shape of the lens
- When light enters the eye it is bent by the lens so that it lands directly on the retina allowing us to see an object
- The closer the object is, the larger the lens becomes and if the object is closer to you then the focal point moves back
- The distance of the object from the lens impacts focal length. The focal length increases as the object move closer to the lens



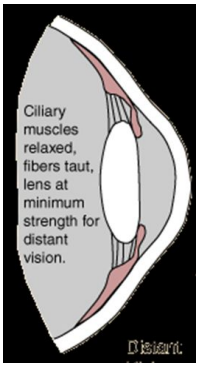
- Light rays from closer objects enter the eye at a greater angle (they are more divergent than light rays from a distant object (they are more parallel)
- Therefore, the rays from a closer object have to be bent more in order for them to be focused at a point on the retina



- The reason you can eventually focus on something close to your eye is that your lens changes shape to accommodate the light
- The focal length of the lens is determined by the shape of the lens, the rounder the lens the more refraction occurs and the shorter the focal length will be
- Accommodation describes the focusing of objects at different distances by changing the shape of the lens and therefore its refractive power
- A flat lens has a lower refractive power
- A round lens has a high refractive power

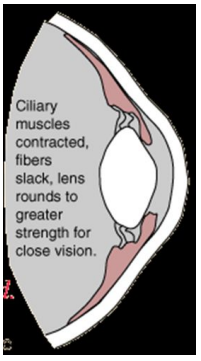
- As humans get older, the ciliary muscles can lose elasticity and there is a tendency for the lens to stay elongated. This can be corrected by glasses

Distant Vision:



- Distant vision means the lens needs to be elongated and relatively flat
- Ciliary muscles are relaxed and hold the suspensory ligaments taut
- Ligaments pull on the lens keeping it flat

Near Vision:



- Lens curvature must increase meaning thicker lens has more refractive power and a shorter focal length
- Ciliary muscles contract meaning suspensory ligaments loosen and the lens becomes rounder

Visual Defects:

- Visual defects can be genetic or environmental
- It can also be caused by aging or a side effect of disease/medication
- It can occur because the lens is damaged or the eyeball is misshaped i.e. not focusing the light on the retina accurately

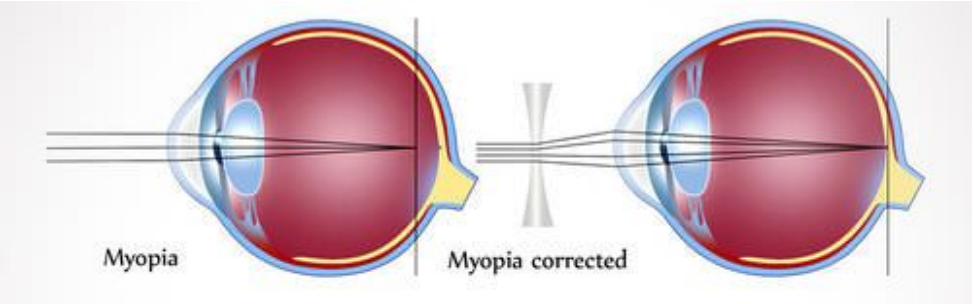
Visual Disorders:

- Astigmatism → refractive error in the eye meaning light doesn't focus on the retina evenly
- Cataracts → clouding of the lens with age, light can't pass through causing cataract blindness
- Colour-blindness → genetic disorder that inhibits functions of cones in the retina (red/green colour blindness most common)
- Macular degeneration → deterioration of the macula of the retina, usually due to old age
- Glaucoma → damage to the optic nerve due to increased pressure within the eye (it can be caused by diabetes)

Myopia - Short Sightedness:

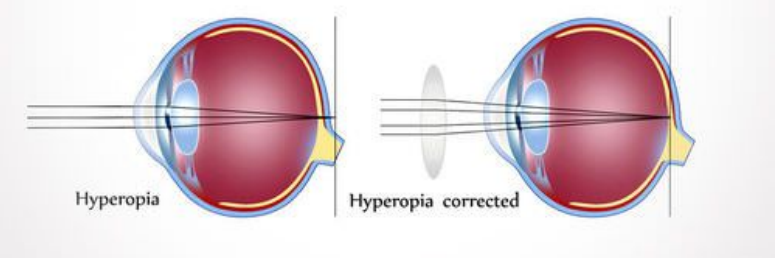
- When distant objects appear blurry
- The focus image (that is far away) is formed in the front of the retina (formed too early)
- It can be caused by an elongated eyeball, the limited refractive power of the cornea or the lens may not flatten enough when ciliary muscles contract
- Closer objects are viewed without difficulty due to the accommodation of the lens
- Fixed by:
  - Glasses
  - Contact lenses
  - Surgery
- Correcting myopia needs a concave lens in spectacles or contact lenses are needed

- Concave lenses cause the light to diverge before it gets to the eye thus allowing the image to fall on the retina rather than in front of it
- By adding the lens, you can see the focal point move to the back of the retina so that image can be formed properly



<u>Hyperopia - Long Sightedness:</u>

- Near objects can not be seen
- The focused image falls behind the retina
- It can be caused by a short eyeball or poor accommodation in the lens (muscles are weaker)
- Glasses or contact lenses containing convex lenses
- This allows the light to converge so that the image falls on the retina not behind it



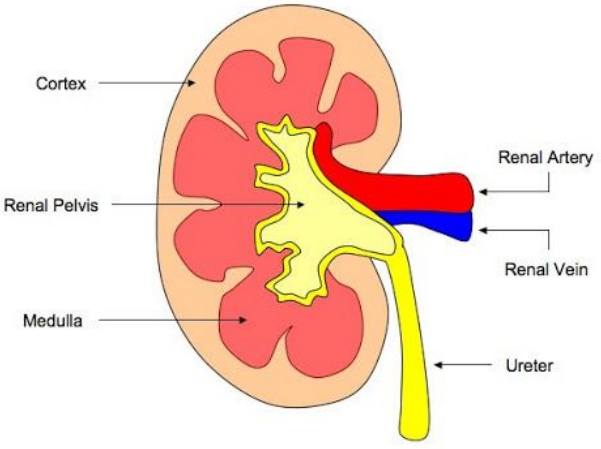
<u>Cataracts:</u>

- Clouding of the lens most commonly due to old age
- Proteins in the lens clump together, preventing light from entering the eye and causing blindness

5.1.3 Loss of Kidney Function

<u>Kidney Anatomy and Function:</u>

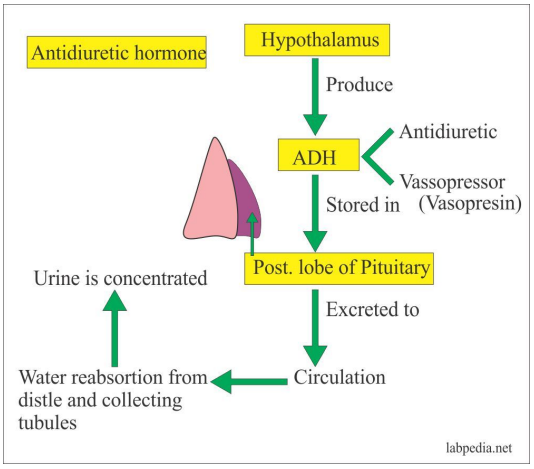
- The kidney is apart of the excretory system and assists in maintaining a balance of water and other fluids
- The importance of waste removal:
  - Wastes are the byproducts of metabolic processes in cells
  - If they accumulate in the body there can be several effects:
    1. Diminished enzyme function
    2. Damage to cellular components
    3. Take up space needed by normal structural and functional chemicals



- Nitrogenous waste:
  - When proteins are metabolised, the byproducts are nitrogen compounds such as ammonia
  - Ammonia is very soluble in water and creates an alkaline solution
  - This causes enzyme function to stop altogether if not removed immediately (the kidney has to get rid of it quickly to ensure proper function)
- There are two main functions of the kidney (in mammals):
  1. Osmoregulation → regulation of water and salt concentrations in the body
  2. Excretion → of urea produced by animals
- The kidney has three main parts:
  - Cortex (outer layer)
  - Medulla
  - Pelvis (drains to ureter)

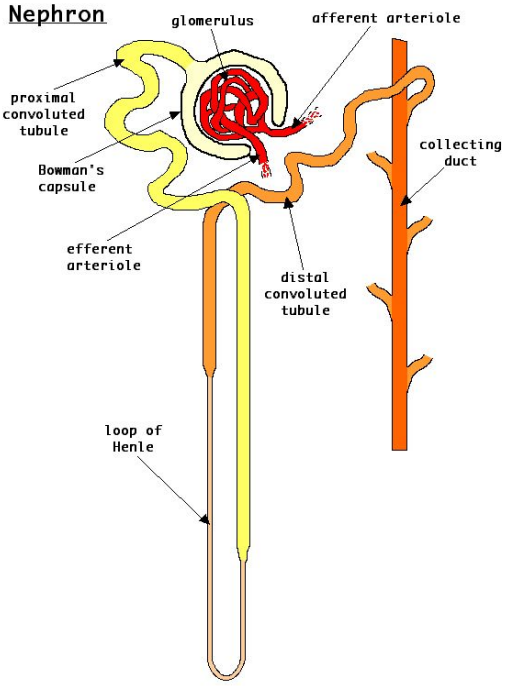
ADH:

- The main action of ADH in the kidney is to regulate the volume and osmolarity of the urine. Specifically, it acts in the distal convoluted tubule (DCT) and collecting ducts (CD). During states of increased plasma osmolality, ADH secretion is increased.
- The antidiuretic hormone stimulates water reabsorption by stimulating insertion of "water channels" or aquaporins into the membranes of kidney tubules. These channels transport solute-free water through tubular cells and back into the blood, leading to a decrease in plasma osmolarity and an increased osmolarity of urine.



The Nephron:

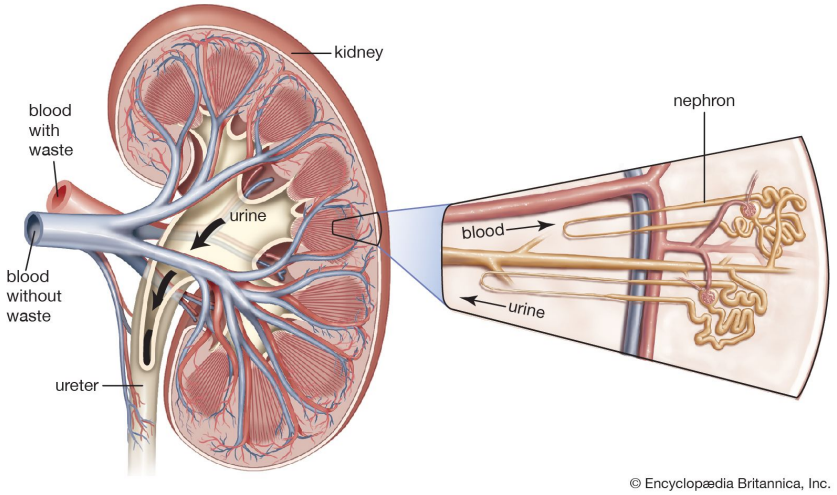
- There are roughly 1,000,000 nephrons per kidney
- They are a significant functional unit of the kidney
- Filtration of blood occurs here
- This results in excretion of nitrogenous waste and osmoregulation (regulation of water and salt)
- Blood and waste enter the kidney via the renal artery
- There are 4 main structures in the Nephron:
  1. Bowman's capsule: It is a cup-shaped structure in the cortex containing the glomerulus
  2. Glomerulus: A network of capillaries, squashed together so that the pressure is high enough to squeeze out the liquid from the blood. Each nephron in your kidneys has a microscopic filter, called a



glomerulus that is constantly filtering your blood. Blood that is about to be filtered enters a glomerulus, which is a tuft of blood capillaries (the smallest of blood vessels).

3. Kidney tubule:
    - The filtrate drains from the Bowman's capsule in the kidney tubule. This is when water and plasma are reabsorbed into the body by passing into capillaries around the tubule
    - The tube then passes up into the cortex again
    - Capillaries surrounding it and salts/glucose are reabsorbed into the blood
    - The kidney tubule travels through both the cortex and the medulla
4. Collecting duct: The functions of the collecting tubes are the transportation of urine and absorption of water. It is thought that the tissue of the kidney's medulla, or inner substance, contains a high concentration of sodium.

- Steps in the Nephron:
    1. The first step in the nephron is filtration (Bowman's capsule and glomerulus):
        - Non-selective
        - Occurs in glomerulus
        - Waste, water and plasma are removed from the blood
    2. The second step in the nephron is reabsorption:
        - Highly selective
        - This occurs in the kidney tubule (medulla)
        - Some plasma and water is reabsorbed by blood (plus some salts and glucose)
        - Passing back into capillaries that surround the tubule (goes back into the bloodstream)
    3. The third step in the nephron is secretion:
        - Selective process
        - Occurs in the kidney tubule
        - Body activity transports substances from blood into the filtrate (this includes wastes)
        - Includes hydrogen, potassium, ammonia and some drugs



Loss of Kidney Function:

- Loss of kidney function can result in a build-up of wastes, electrolytes and dangerous levels of fluid in the blood
- Symptoms often appear very late in the disease and there can be a 90% loss in kidney function before symptoms appear
- Kidney failure occurs due to many reasons:
  - Diabetes
  - High blood pressure
  - Inflammation
  - Obstruction (kidney stones)
  - Infections
- EXAMPLE → Diabetic kidney disease:
  - Diabetic nephropathy is a common complication of type 1 and type 2 diabetes.
  - Over time, poorly controlled diabetes can cause damage to blood vessel clusters in your kidneys that filter waste from your blood.
  - This can lead to kidney damage and cause high blood pressure.
  - High blood pressure can cause further kidney damage by increasing the pressure in the delicate filtering system of the kidneys.
- Albuminuria is a sign of kidney disease and means that you have too much albumin in your urine. Albumin is a protein found in the blood. A healthy kidney doesn't let albumin pass from the blood into the urine. A damaged kidney lets some albumin pass into the urine

5.2 Technological Developments

- *Investigate technologies that are used to assist with the effects of a disorder, including:*
  - *Hearing loss: cochlear implants, bone conduction implants, hearing aids*
  - *Visual disorders: spectacles, laser surgery*
  - *Loss of kidney function: dialysis*

5.2.1 Hearing Loss

Hearing Aids:

- Hearing aids are electronic devices that can amplify sounds entering the external ear and are worn behind the ear or in the ear canal
- They have a microphone to detect and convert sound into electrical signals, an amplifier to strengthen these signals, a receiver to convert the signals back tou sound and a speaker to direct the sound into the external ear
- Hearing aids rely on the natural functions of the ear to perceive the sound and to detect the larger vibrations
- Technological advances have significantly improved hearing aids in recent years meaning that there is no longer excessive background noise

Cochlear Implant:

- There is an external speech processor which captures the sound and transmits it as a digital signal to the receiver
- It is then converted to an electoral signal and passed onto the electrode array in the cochlear which stimulates nerve endings. This is then sent to the brain for processing.
- They are used to treat severe to profoundly death people with missing or damaged hair cells in the cochlea.
- They are worn externally behind the ear and the speech processor detects sounds and converts it into digital code into electrical impulses and sends them along electrodes into the cochlea

Bone conduction implants:

- People with outer or middle ear problems that block or restrict the flow of sound waves can be helped to hear with bone conduction implants.
- An external sound processor is attached either magnetically or directly to a small titanium plant that is surgically placed in the bone behind the ear
- The sound processor detects and converts sounds into vibrations that are transferred directly through bone to the cochlear
- Hair cells within the cochlea convert these vibrations into nerve impulses to be sent to the brain, so that the person can hear
- People with hearing loss can trial bone donation treatment prior to surgery, by wearing the sound processor and external vibraot on the headband

5.2.2 Visual Disorders

- Without management of visual disorders, people can have trouble completing everyday activities and tasks, making it difficult for them to be independent
- There are technologies to assist with these and other visual defects are therefore an advantage to both the individual and society as a whole

Spectacles:

- Glasses correct vision by changing the angle at which light hits the cornea, adjusting for misshapen corneas which cause the focal point to deviate from the norm
- This allows the eye to focus light in the right spot in the eye, on the retina
- Glasses are available with a prescription which means that they are made for each person depending on their needs
- For short-sightedness where the focal point falls in front of the retina, concave lenses allow light to focus on the retina
- For far-sightedness where the focal point falls behind the retina, convex lenses allow light to focus properly on the retina
- Myopia
    - Corrected with spectacles that have a concave lens (thicker towards the outside and thinner towards the centre).
    - These lenses bend the light rays, allowing the focused image of a distant object to fall on the retina instead of in front of it

- Hyperopia:
  - Corrected with spectacles that have a **convex** lens (thicker towards the centre and thinner towards the edges)
  - This type of lens bend incoming light rays inwards, causing them to begin converging before they reach the eye, shortening the focal length and allowing the focused image of a near object to fall on the retina instead of behind it

Laser Surgery:

- Use of lasers to change the curvature of the cornea to alter its refractive power, therefore overriding any problems with the shape of the eye
- This can be used to correct myopia, hyperopia, cataracts and astigmatism
- LASIK:
  - Involves two lasers
  - The first creates a cut/flap in the cornea
  - The second laser is a very high precision laster that shaves of layers of cells from the cornea and adjusts its refractive power down to the microscopic level
  - There are usually eligibility requirements and it is significantly expensive

Cataract Surgery - Phacoemulsification:

- Restores sight by replacing the lens with an artificial one (intraocular lens implantation - IOL)
- It is quick and relatively painless
- The article lens only costs $10 to make but the surgery itself is expensive (surgeons, tools)
- Fred Hollows foundation has had a very significant impact on restoring sight
- During surgery, an incision is made in the cornea, the cloudy cataract lens is removed using high-frequency ultrasound waves and vacuumed up
- The IOL replaces the lens restoring sight

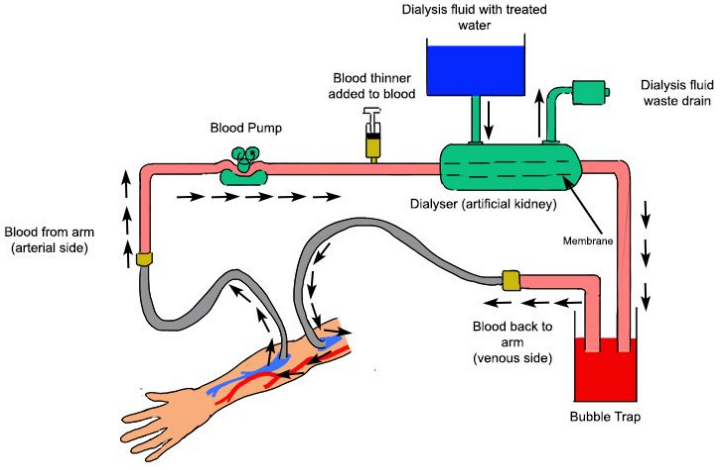
5.2.3 Loss of Kidney Function

Dialysis:

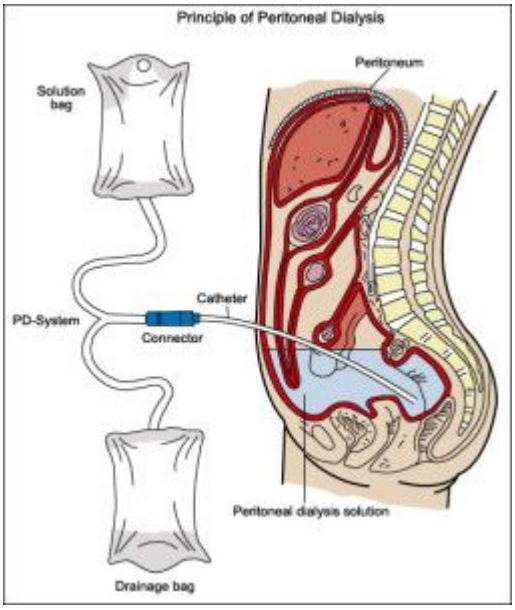
- A process that artificially removes salt and extra water from the blood across a semipermeable membrane
- Dialysis is needed when kidneys are failing to adequately filter the blood
- Dialysis is not a cure for kidney failure but instead an effective treatment
- There are various types of dialysis including:
  - Haemodialysis
  - Peritoneal dialysis

Haemodialysis:

- This uses a dialysis machine to filter the blood
- A needle (called a cannula) is placed in the arm



- Blood flows out into the machine and through a series of semi-permeable membranes
- Toxins are removed and clean blood is returned to the body
- Can be done at home but requires specialist equipment or the patient will have to travel regularly to a medical centre
- Have to undergo this process 3 times a week and each session can take up to 3-5 hours. This means that affected individuals are several sontrainted in terms of working and traveling ability
- The process is also expensive and uncomfortable



Peritoneal Dialysis:

- It is a way to remove waste products from your blood when your kidneys can't adequately do the job any longer. This procedure filters the blood in a different way (compared to hemodialysis)
- A catheter is placed into the abdomen and a cleansing fluid is pushed into the cavity
- The wastes then filter from the blood into the fluid in the cavity
- This fluid is removed from the abdomen and discarded
- Must be repeated multiple times during the day
- There is a risk of infection due to the catheter tubing
- When you start treatment, dialysis solution—water with salt and other additives—flows from a bag through the catheter into your belly. When the bag is empty, you disconnect it and place a cap on your catheter so you can move around and do your normal activities. While the dialysis solution is inside your belly, it absorbs wastes and extra fluid from your body.

5.3 Evaluation of Technologies

- *Evaluate the effectiveness of a technology that is used to manage and assist with the effects of a disorder*

Known the benefits and limitations, how does it benefit the individual and society by restoring their sight (improves quality of life). Technology is a great benefit

5.3.1 The Ear - Evaluation

Cochlear Implant:

- A cochlear implant is a surgically implanted neuroprosthetic device to provide a person with moderate to profound sensorineural hearing loss a modified sense of sound.
- CI bypasses the normal acoustic hearing process to replace it with electric signals which directly stimulate the auditory nerve.

Advantages	Disadvantages
- People are able to better communicate, watch TV, listen to music, make phone calls and it can generally improve quality of life. Communicating with people is a massive part of being a human and therefore necessary - It is a very successful surgery and only has a fail rate of 0.2% - It usually lasts an entire lifetime (only occasionally needs replacement/repair) - Ultimately assists with deafness and drastically improves the quality of life	- There can sometimes be complications involved in the process and people can experience: dizziness, vertigo, injury to the facial nerve (muscles are ruined), tinnitus (a ringing or buzzing sound in the ear), potential taste disturbances and meningitis (an infection of the lining of the surface of the brain) - The quality of sound and sometimes described as mechanical or artificial. This means it is 'not worth it' for some people. - The implants can fail completely - They are also extremely expensive and can cost around $25,000 – $50,000

5.3.2 The Eye - Evaluation

LASIK (Laser eye surgery):

- Considered an elective surgery to fix long and short-sightedness
- A type of refractive surgery for the correction of myopia, hyperopia, and astigmatism.
- Laser eye surgery uses a specialized laser to ablate the cornea, reshaping it to correct refractive errors. In the case of hyperopia, the laser shapes the cornea to make it steeper, lengthening it so the focusing power is directed onto the retina instead of behind it.
- Laser eye surgery. Laser eye surgery involves using a laser to burn away small sections of your cornea to correct the curvature so light is better focused onto your retina. Instead of falling short of the retina, the shape of the cornea now allows it to land directly on the retina.

Advantages:	Disadvantages:
- The procedure only takes 20-30 minutes and recovery is relatively quick - No need to purchase/wear glasses - Needed for occupation, therefore, correcting vision is an advantage - No bandages or stitches are required - Approximately 85% of people return to 20/20 vision (more independence, can do the job)	- Costs $2500-$4000 per eye - Isn't covered by medicare since it is elective surgery. Might be protected by private health insurance. - Only for 18 years and above - Risks and complications are involved: could over/under correction, eye infection, dry eyes

5.3.3 The Kidney - Evaluation

Hemodialysis:

- Hemodialysis is a treatment to filter wastes and water from your blood, as your kidneys did when they were healthy.
- Hemodialysis helps control blood pressure and balance important minerals, such as potassium, sodium, and calcium, in your blood.
- Needed when kidneys can no longer remove enough wastes and fluid from the blood. Dialysis is needed when an individual only have 10% of their kidney function left
- Hemodialysis is a procedure where a dialysis machine and a special filter called an artificial kidney, or a dialyzer, are used to clean blood. For blood to get into the dialyzer, the doctor needs to make an access, or entrance, into blood vessels. This is done with minor surgery, usually in the patient's arm.
- For example, peritoneal dialysis may be recommended for:
    - Children aged 2 or younger
    - People who still have some limited kidney function
    - Adults who do not have other serious health conditions, such as heart disease or cancer

Advantages	Disadvantages
- It can increase the quality and quantity of life - It can occur within the person's home meaning it is more convenient	- Very time consuming as it has to occur 3 times per week and can take up to 4 hours at a time. This means limited travel and work opportunities - It can be a very uncomfortable process and people can also experience headaches and cramps due to the procedure - It is a very expensive process - There can also be infection involved