

HEREDITY

REPRODUCTION

INQUIRY QUESTION: How does reproduction ensure the continuity of a species?

1. THE MECHANISMS OF REPRODUCTION THAT ENSURE THE CONTINUITY OF A SPECIES

⇒ 1.1 Sexual and asexual methods of reproduction in a variety of organisms

- Reproduction is fundamental to the continuation of life
- ASEXUAL is the production of offspring that are genetically identical to each other and the parent
- SEXUAL is the production of genetically different offspring resulting from two parents, each producing gamete which fuse in a process called fertilisation

SEXUAL REPRODUCTION	ASEXUAL REPRODUCTION
An essential step is cell division by meiosis	Processes include binary fission and/or mitosis cell division
Requires two parents of different gender Requires fertilisation, which results in offspring with a new, unique combination of genetic material, some from each parent	Results in offspring that are genetically identical to the parent
Fertilisation can occur internally or externally	
Dominant form of reproduction in many multicellular plants and animals	Dominant form of reproduction in many unicellular, colonial or simple multicellular organisms
Animal examples include egg laying in insects, crustaceans, arachnids, amphibians, fish, birds, reptiles and monotremes; live birth in placentals;	Examples include spore production in fungi, and budding and binary fission in bacteria or protists

immature birth. And pouch-raising in marsupials

Plant examples include production of flowers or cones

Plant examples include production of bulbs, tubers, rhizomes, stolons and corms

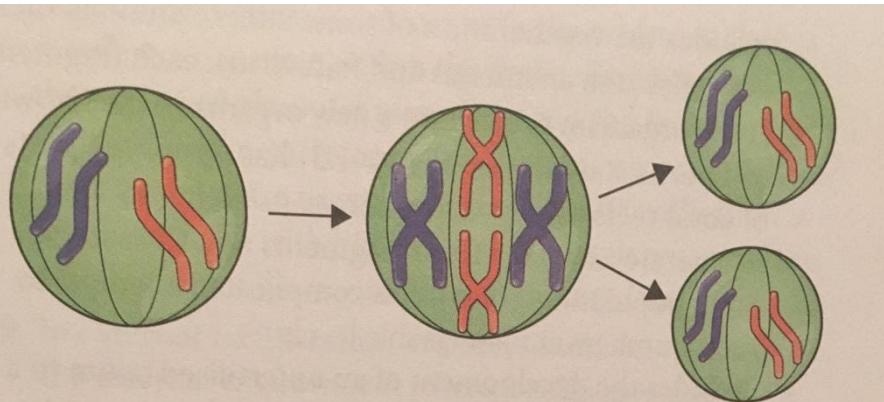


Figure 1.1 Summary of mitosis in a cell in which $2n = 4$. It results in two daughter cells identical to the eukaryotic parent cell.

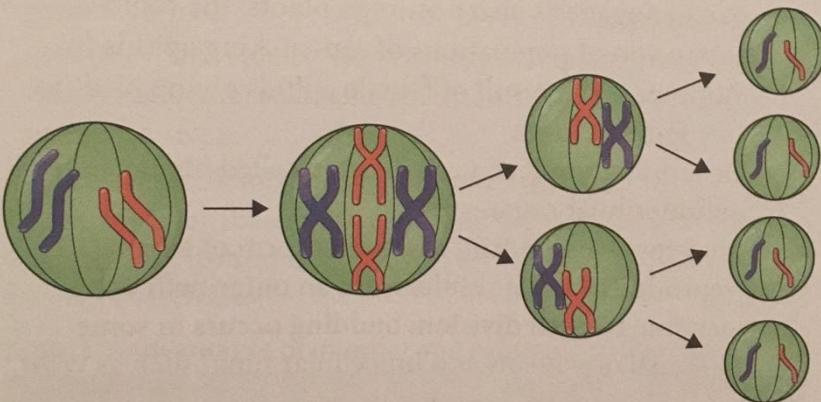


Figure 1.2 Summary of meiosis in a cell in which $2n = 4$. It results in four unique daughter cells, each with n chromosomes (i.e. one member of each homologous chromosome pair).

○ Meiosis is a cellular reproductive process that occurs in eukaryotic cells; meiosis results in the formation of gametes that contain half the normal complement of genetic material

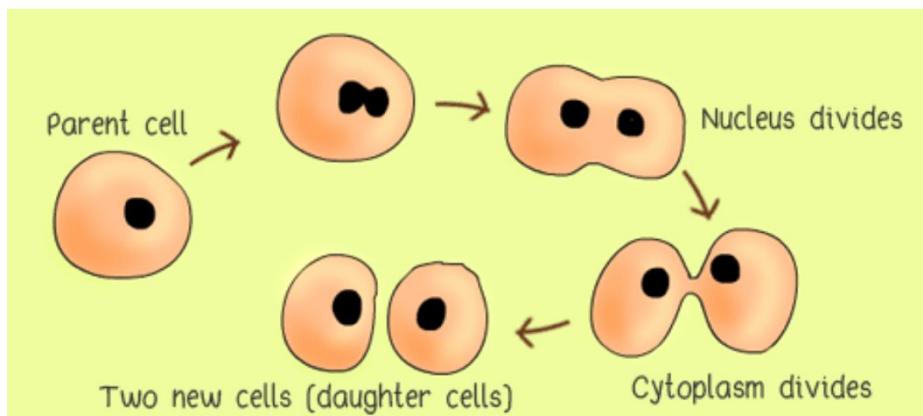
○ Mitosis is a cellular reproductive process that occurs in eukaryotic cells, mitosis refers to the replication and division of the nucleus; mitosis results in the production of two identical daughter cells from the parent cell; cytokinesis, which follows mitosis, results in the formation of two cells with each containing a nucleus and cytoplasm

ASEXUAL REPRODUCTION

○ Asexual reproduction:

- Occurs widely in plants, generally in the production of runners, rhizomes, tubers, bulbs, corms and spores
- Results in clones of the original parent
- Includes the regeneration of some simple animals (starfish, flatworms); fragments of organism grow into a new organism
- Includes development of an unfertilised ovum in a process called parthenogenesis in some species (shrimp, bees, ants)

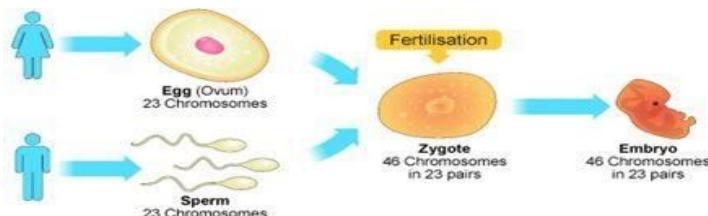
- Occurs as a result of a cell division called binary fission in unicellular organisms
- May involve budding as another form of asexual reproduction that results from an outgrowth or asymmetric cell division
- Advantages of asexual reproduction
 - Only one parent is needed
 - Rapid population growth of identical members
- Disadvantages of asexual reproduction
 - Lack of biodiversity within a population
 - Overpopulation, leading to possible negative ecological impacts



SEXUAL REPRODUCTION

- Sexual reproduction
 - Occurs widely in animals and seed-bearing plants; angiosperms (flowering) and gymnosperms (cone-bearing)
 - Occurs in primitive plants such as ferns, mosses and liverworts
 - Fungi may reproduce sexually in times of bad conditions
 - Results from the production of two different types of gametes (male and female)
 - Male gamete is smaller and more mobile - female is larger containing food reserves for the early development of the zygote into an embryo
 - Female gamete develops into the seed or egg of embryo which becomes a foetus
 - Comes about due to male and female gametes uniting in a process called fertilisation
 - Fertilisation results in a zygote which grows through mitotic cell divisions and differentiation to produce an embryo
 - Results from organisms of different gender, population growth rates are often linked to the proportion of females , as female reproductive cycles become the limiting factor

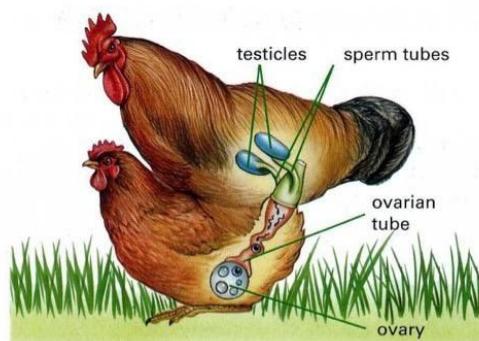
- Occurs in eukaryotes. As the genetic material is carried on chromosomes the gametes have half of the normal number of chromosomes
- Somatic cells are diploid
- Advantages of sexual reproduction
 - Biodiversity or offspring
 - Variation in offspring is the basis of evolution by natural selection
 - Methods of sexual reproduction promote the dispersal of offspring in some species
- Disadvantages of sexual reproduction
 - Generally requires two parents of different sex to produce viable gametes and fertilisation
 - Requires fertilisation



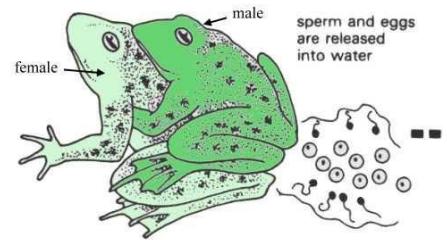
Sexual Reproduction

⇒ 1.2 Advantages of external and internal fertilisation in animals

- In sexual reproduction fertilisation is either internal or external
- INTERNAL FERTILISATION occurs with the release of the motile male gamete. Within the female body such as in the vagina of placental mammals, and the fertilisation of the female gamete. Internal fertilisation ensures that fewer male gametes are unproductive. Internal fertilisation is also associated with higher levels of development of the embryo internally or under parental care
 - The union of male and female gametes that occur within the body of the female; in mammals this results from copulation (sex) involving penetration of the female by the penis of the male and release of semen



- EXTERNAL FERTILISATION ova and sperm are released into the external environment where fertilisation and subsequent development may occur
 - The unions of male and female gametes after their release outside the body, often in an aquatic environment



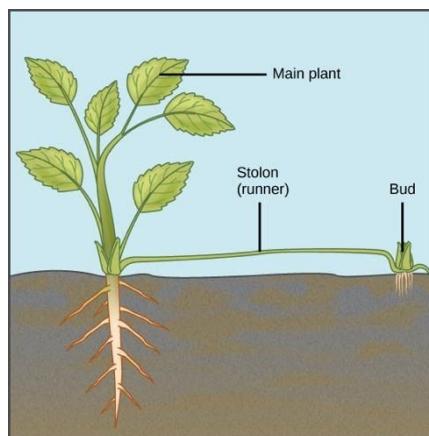
INTERNAL FERTILISATION	EXTERNAL FERTILISATION
Gametes, zygote and embryo contained in a protected environment: controlled temperature, moisture, free from predators	Gametes, zygote and embryo are usually in an aqueous environment for ready dispersal
Relatively high success in development of offspring due to increased probability of fertilisation occurring and many instances of paternal care of offspring; less need for production. Of large numbers of gametes	Development of offspring independent of parents; parents can put energy into the production of large numbers of gametes
Well suited to a range of terrestrial and aquatic environments	Well suited to moist or aquatic environments

⇒ 1.3 Asexual and sexual reproduction in plants

- Reproduction in terrestrial plants is both sexual and asexual
- Most terrestrial plants are sessile, so successful reproduction is a means of competition and means of dispersal

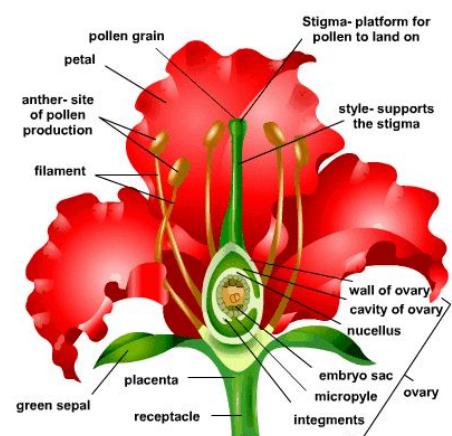
ASEXUAL

- Involves mitotic cell division in specific locations such as buds
- New individuals arise from portions of the roots or stems, leaves or buds and are genetically identical to the parents
- Adult plants produce vegetative organs such as bulb, tubers, rhizomes and suckers from which new plants can arise
- Known as vegetative propagation
- It is a form of cloning; humans use it to have desirable characteristics



SEXUAL

- Mainly occurs in angiosperms and gymnosperms
- They produce the seeds that contain the embryo
- Great variety of ways male gamete is transferred to the female gamete in a process called pollination
- Angiosperms have ovules protected within the ovary
- Gymnosperms have seeds that are not protected in an ovary
- Seeds produced by angiosperms and gymnosperms contain food reserves for the germinating embryo



STAMEN	CARPEL
<p>Anther – where pollen grains are formed</p> <p>Filament – stalk that carries the anther. The length determines whether the anthers are contained inside the petals for insect pollination or outside for wind pollination</p>	<p>Stigma – sticky top surface to which pollen adheres. May be relatively small and smooth (in insect-pollinated plants) or large and feathered (wind)</p> <p>Style – joins stigma to ovary</p> <p>Ovary – where ovules are formed</p>

Petals – a whorl of leaves modified to increase the likelihood of pollination. Often brightly coloured and scented to attract pollinators. May have complete shapes to facilitate entry of particular pollinators to the flower.

Receptible – reinforced base of the flower, which supports the weight of the reproductive structures

Sepals – a whorl of modified leaves protecting the unopened bud

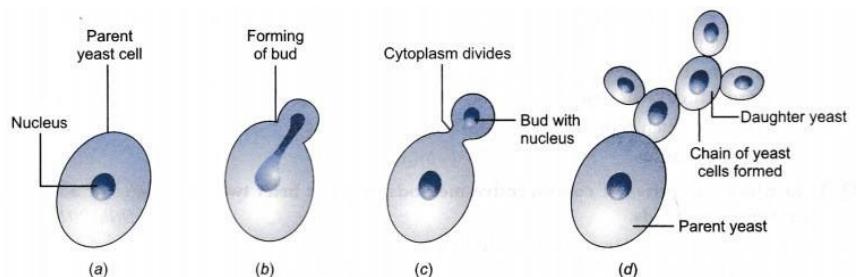
- Male. Gametes inside the pollen must be carried from the anthers to the stigma a.k.a POLLINATION
- Pollen tube germinates and grows down the style carrying sperm to the ovary/ovule

⇒ 1.4 Fungi: budding and spores

- Fungi include yeasts, mushrooms, moulds, mildew, rust and smut
- Reproduction is asexual and occurs through budding, spore production and fragmentation (fragment of organism detaches and grows into independent organism)

BUDDING

- Adult organism gives rise to a small bud which separates to grow into a new individual
- Small outgrowth occurs on the bud, parent replicates DNA, nucleus splits
- Good if there is no environmental change
- Bad if environment changes, no variation, extinction



SPORES

- Spores are tiny unicellular reproductive cells
- Light and can be carried far distances by wind or animal, colonize new environments
- Able to reproduce rapidly and colonise a wide area ensuring continuity of a species

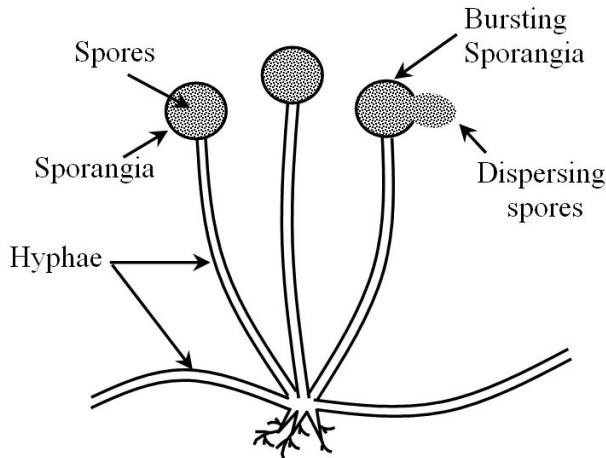
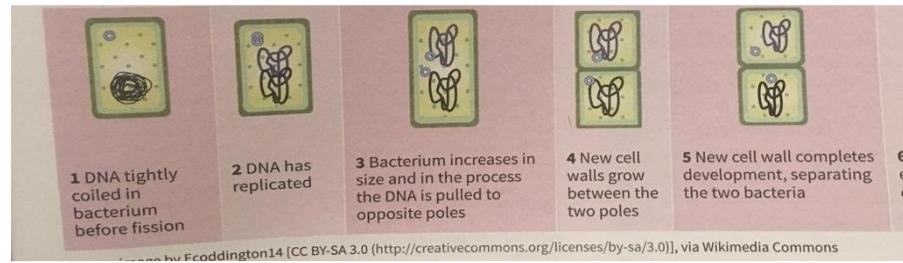


Fig. 5 Spore formation in Rhizopus

⇒ 1.5 Bacteria: binary fission

- Bacteria are prokaryotes, duplication of DNA and cell division varies from mitotic divisions in eukaryotic cells
- Comes in a wide range of types and roles
- Reproduce asexually through binary fission or multiple fission
- Transfer genetic material in the form of a plasmid or a small circular piece of DNA
- Bacteria double every 20 minutes
- Binary fission is a form of mitosis that is used by unicellular organisms such as bacteria. The process starts with the copying



the genetic material (in the form of bacterial chromosomes) of the parent cell. Each chromosome moves to each side of the cell. This is followed by the elongation of the cell and cytokinesis which is the splitting of the cell membrane and cytoplasm of the cell into two daughter cells. As there is no cell nucleus in bacteria, there will not be the splitting of cell nucleus. It is important to note that the parent cell won't exist at the end because it is now part of the two daughter cells. The two daughter cells are genetically identical to each other as well as identical to the parent which they obtained their genetic information came from.

⇒ 1.6 Protists: binary fission and budding

- Reproduce through binary fission and budding
- When budding occurs offspring may stay attached to parent resulting in a colony
- Budding in protists starts off by the parent protozoan producing a bud which is a daughter nucleus that is created based on the separation of the parent protozoan's cytoplasm (one fission).
- The mechanism of binary fission in protist is similar to that of bacteria's binary fission process. However, as DNA is stored in the nucleus (whereas no nucleus in bacteria), the chromosome will move to each side of the nucleus before the splitting of the nucleus and eventually splitting of the cell membrane and cytoplasm into two daughter cells.

2. FERTILISATION, IMPLANTATION AND HORMONAL CONTROL OF PREGNANCY AND BIRTH IN MAMMALS

⇒ 2.1 Fertilisation in mammals

- Copulation (sex) may result in internal fertilisation
- Males release semen containing spermatozoa from the penis in a process called ejaculation into the urogenital sinus of monotremes and marsupials and into the vagina of mammals
- Spermatozoa travels to the oviducts which may contain an oocyte that is released from a follicle in the female ovary
- 200-500 million spermatozoa reach the oocyte
- Testes produce haploid spermatozoon through meiosis of the spermatocyte. It consists of:
 - A head which contains DNA
 - A neck which contains centrioles that support zygote division
 - Mitochondria to supply energy

- Flagellated tail that helps sperm swim

Semen: fluid containing spermatozoa and other secretions for transport

Spermatozoa: male gametes; small motile cells carrying a haploid set of chromosomes, produced in the testes

Ejaculation: process of releasing semen from an erect penis

Oocyte: immature female germ cells that gives rise to the ovum of female gamete containing a haploid set of chromosomes

- o Ovary contains follicles in which meiosis occurs to produce the oocyte
 - Process occurs in two stages
 - 1. In female foetus
 - 2. Occurs at intervals after puberty when the ovarian follicle ruptures to release the oocyte
 - Follicle becomes the corpus luteum which plays a role in pregnancy
 - Meiosis is complete after the entry of a spermatozoa
 - The oocyte is then called the ovum
- o Ovum in placental animals is 0.07-0.15 mm
- o Oocyte has two external layers;
 - Inner jelly coat
 - Outer layer of cells that are remnants of the follicle
- o Spermatozoa binds with receptors on the jelly coat; acrosome releases enzymes that help it penetrate the layer and change its nature
 - Ensures only sperm of the same species can enter
- o Two plasma membranes fuse, allowing the spermatozoa to enter the cytoplasm of the oocyte
 - Causes the cortical granules to release their contents to further ensure only spermatozoa enters the cell
- o Oocyte has suspended meiosis
 - Entry of sperm triggers the oocyte to complete its meiotic cell division
 - Now called an ovum as it contains a pronucleus
 - Membranes of two nuclei dissipate and the two sets of haploid chromosomes join together
 - Pronuclei fuse, the diploid nucleus of the zygote has formed, and fertilisation is complete

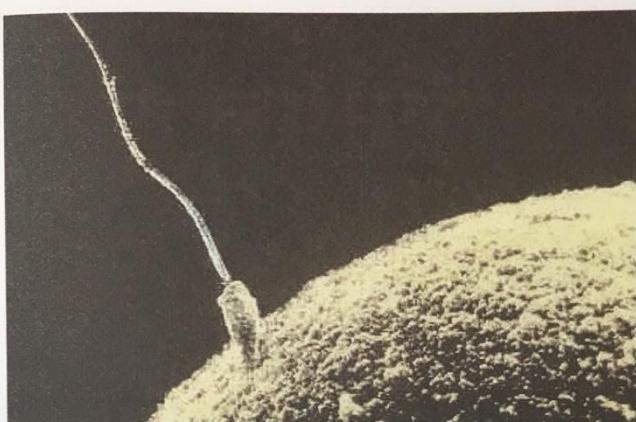
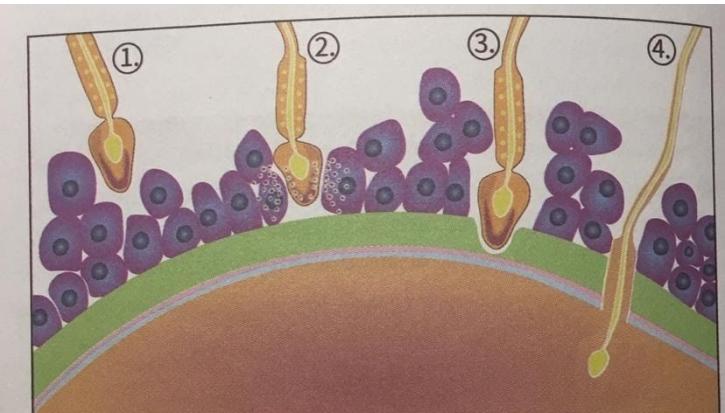


Figure 1.15 Scanning electron micrograph of spermatozoon about to penetrate the oocyte



1. Spermatozoon approaches oocyte.
2. Spermatozoon releases enzymes to assist penetration of the outer layer of the oocyte.
3. The inner jelly coat changes structure in many mammals, preventing the entry of further spermatozoa.

⇒ 2.2 Implantation in mammals

- After fertilisation, zygote starts a process of mitotic cell divisions to become a solid ball of cells called a morula
- Morula travels down the oviduct
- It divides in the uterus and differentiates into the hollow fluid-filled ball of cells called the blastocyst. This embeds (implants) into the endometrium

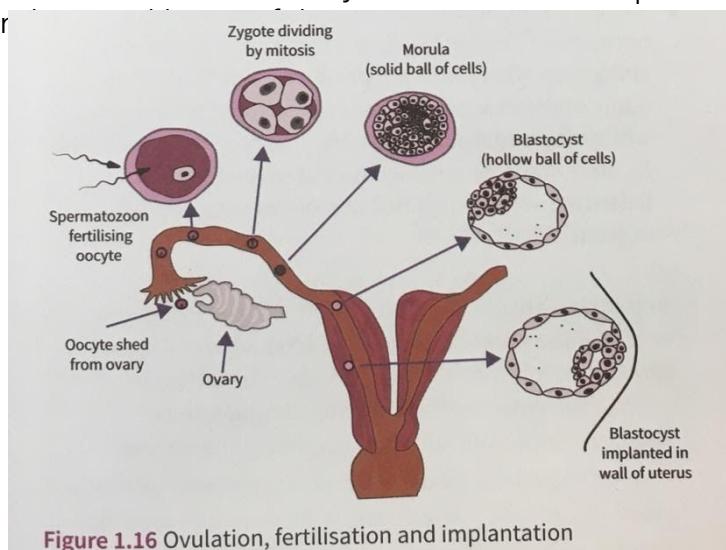


Figure 1.16 Ovulation, fertilisation and implantation

- Jelly coat is shed prior to implantation
- Implantation is a complex process
- Inner cell mass contains stem cells that will differentiate into the embryo, a cavity and an outer cell layer called the trophoblast
- Occurs in several ways
 - HUMANS trophoblast cells produce and release protein-dissolving enzymes that assist the erosion of the endometrium
 - Process assists the embedding of finger-like projections into the uterus wall
 - Enzymes assist by digesting some cells in the endometrium that become the source of nutrition for the developing embryo

- After blastocyst has implanted, stem cells start differentiating and the body plan of the new embryo starts to develop
- After implantation in placental mammals the trophoblast develops into the placenta

⇒ 2.3 Hormonal control of pregnancy in mammals

- In mammals' hormones are released into the blood by endocrine glands.
 - Hormones promote changes in cells and tissues such as increased growth, development of secondary sexual characteristics and control of the menstrual cycle in female primates and the oestrous cycles in other groups of female mammals
- Placental mammals require a complex hormonal coordination of ensure the development of embryo and foetus
 - Blastocyst produces CG (chorionic gonadotrophin) which prevents corpus luteum from degenerating
 - Corpus luteum is the remnant of the ovarian follicle that released the oocyte and it produces hormones that maintain pregnancy

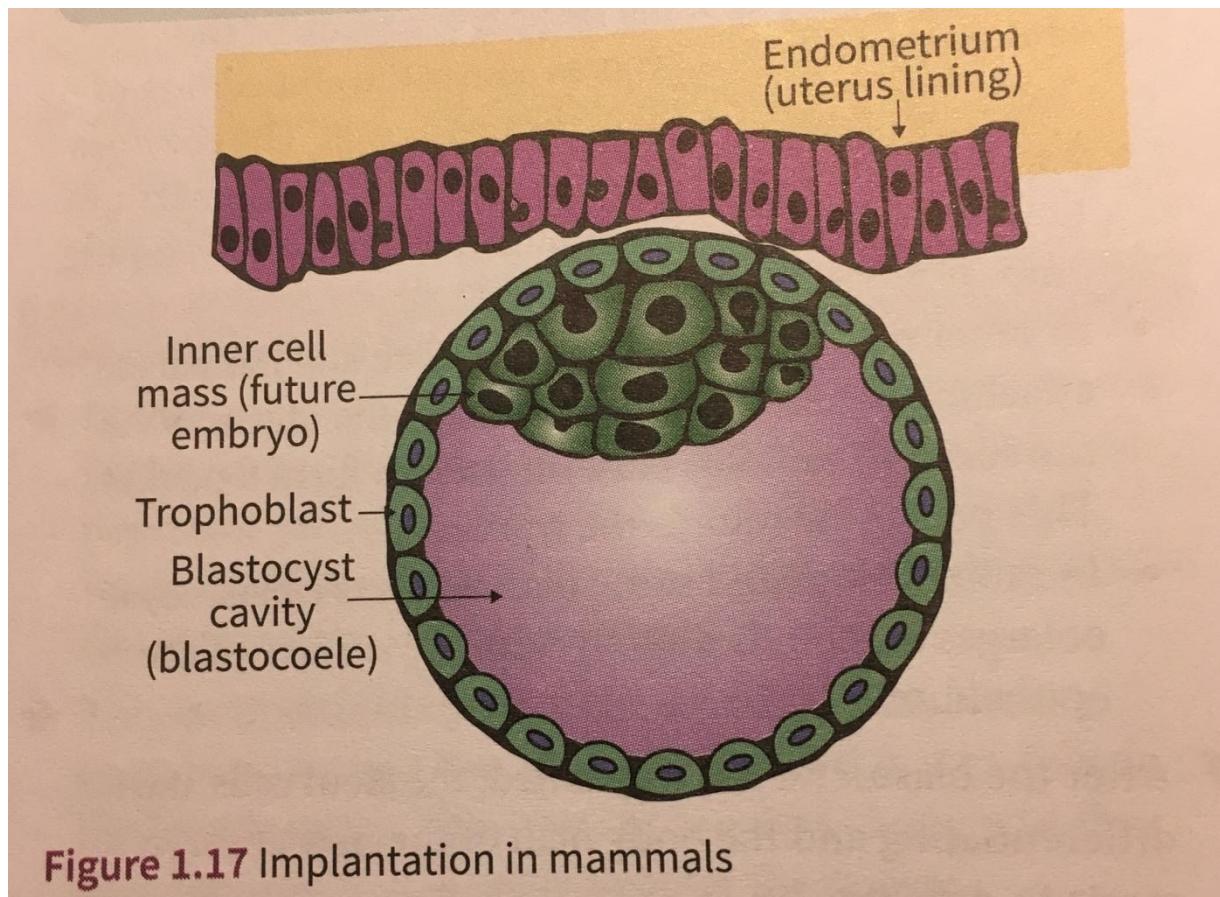


Figure 1.17 Implantation in mammals

HORMONE	WHERE PRODUCES	ROLE
Progesterone	<p>Placenta and corpus luteum</p> <p>In human, horses, sheep and cats the progesterone released from the placenta is sufficient to maintain pregnancy</p> <p>Cattle, pigs, dogs and goats rely heavily on the progesterone released from the corpus luteum</p>	<p>Called the hormone of pregnancy because without it the endometrium would not be maintained or support the implantation of an embryo</p> <p>Stops contraction of the smooth muscle of the uterus which is involved in giving birth; if occurring sufficiently before full gestation this may result in a miscarriage</p> <p>Prevents the pituitary secreting gonadotrophin luteinising hormone and follicle stimulating hormone to suppress ovulation during pregnancy</p> <p>Helps induce immune tolerance, possibly mediated by a protein called Progesterone Induced Blocking Factor (PIBF)</p>
Oestrogens	<p>Placenta, E4 from foetal liver</p>	<p>Stimulates the development of mammary glands</p> <p>Stimulate the development of the uterine smooth muscle layers ready for birth; this counters the effect of the 'progesterone block' that suppresses uterine contractions late in pregnancy</p> <p>Helps induce immune tolerance, possible by expanding regulatory B and regulatory T or Tregs cells o</p> <p>Oestriol is made from a precursor produced in the adrenal glands of the foetus; its levels late in pregnancy are considered an accurate indicator of the health of the foetus</p>

		<p>E3 promotes uterine growth and sensitivity to other pregnancy hormones and is believed to trigger labour when it becomes the dominate hormone</p> <p>E4 is believed to protect the foetus from maternal oestrogen</p>
Human Chorionic Gonadotrophin (hCG)	Blastocyst	<p>Prolongs the activity of the corpus luteum</p> <p>Helps induce immune tolerance, possibly expanding regulatory B and regulatory T</p> <p>Induces production of relaxin and stimulates production of oestrogen and progesterone</p> <p>Its presence in urine or blood indicates pregnancy has occurred</p>
Relaxin	Corpus luteum of ovary and placenta	<p>Relaxes maternal muscle joints and ligaments to allow for the expanding foetus</p> <p>Relaxes uterine muscles and prepares lining for implantation</p>
Corticotrophin-releasing Hormone (CRH)	Hypothalamus of brain and foetus and placenta during pregnancy	<p>Increases the production of the stress hormone cortisol</p> <p>Regulates duration of pregnancy and foetal maturation</p> <p>Suppresses maternal immune response in early pregnancy</p> <p>Improves blood flow between placenta and foetus inn later pregnancy</p> <p>Associated with 'late term cortisol surge' that is believed to support maternal bonding and care in humans and some animals</p>
Placental Lactogen or Chorionic Somatomammotropin	Placenta	Possible controls metabolism of foetus and mother, mobilising energy reserves for the foetus

		Stimulates corpus luteum function
		Promotes development of mammary glands before birth

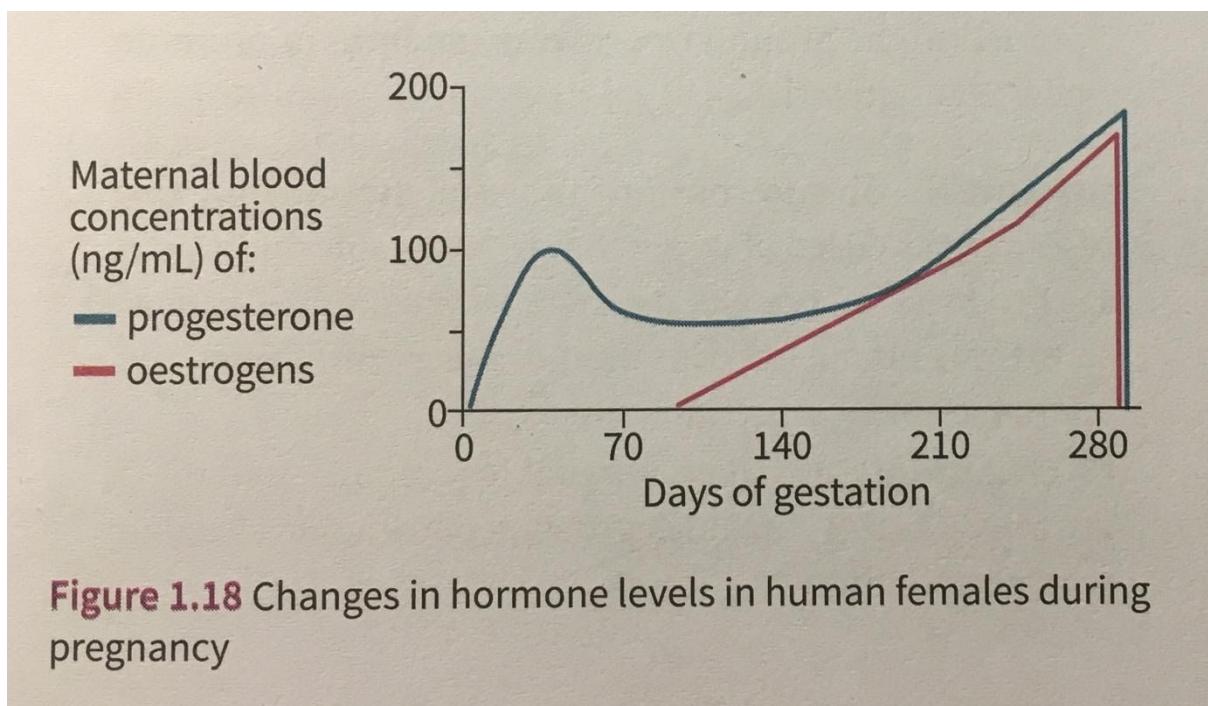


Figure 1.18 Changes in hormone levels in human females during pregnancy

⇒ 2.4 Hormonal control of birth in mammals

- In placental mammals, birth is the process in which the foetus is delivered from the uterus to the outside world
- Involves contractions of the uterine smooth muscles, widening and dilation of the cervix and the delivery of the foetus through the cervix and vagina
- Birth is orchestrated by a number of interacting hormones which may coordinate for the same role or balance each other in contradictory roles until one becomes dominant
- Birth is generally followed by the expulsion of the placenta

HORMONE	WHERE PRODUCED	ROLE
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Corticotrophin-releasing hormone (CRH) and cortisol (stress hormone)	Foetus and uterus	Believed to trigger a rise in oestriol and when this hormone becomes dominant, birth ensues; these stress hormones are the result of the increased pressure due to the growing foetus and their levels rise just before birth
Oestriol (E3) – an oestrogen associated with birth	Placenta	Counters the role of progesterone on suppressing uterine contractions increases sensitivity of uterus to other hormones
Oxytocin	Hypothalamus and stored and released from posterior pituitary; also released from foetus	Has a major role in birth as well as other functions such as ejection of milk while breastfeeding and maternal bonding Initiates and strengthens rhythmic uterine contractions
Prostaglandins	Uterus	Helps initiate labour and uterine contractions by reducing progesterone (which inhibits uterine contractions)
Relaxin	Ovaries	Helps dilate (open) the cervix to allow passage of foetus; widens pubic bone and relaxes pelvic ligaments

3. THE IMPACT OF SCIENTIFIC KNOWLEDGE ON THE MANIPULATION OF PLANT AND ANIMAL REPRODUCTION IN AGRICULTURE

- Human population increases has increased the demands on agricultural production

⇒ 3.1 Impact of scientific knowledge on the manipulation of plant reproduction in agriculture

ASEXUAL TECHNIQUES

- Use of cloning plants through vegetative propagation which allows rapid development of plants with known qualities
- Plants often are able to regenerate after damage
 - Cutting a section of stem, leaf, root and treating it with plant growth hormones to stimulate the development of roots or mitosis and then planting these 'cuttings', results in plants identical to the parent
- Tissue culture uses knowledge of where cell division occurs in plants to produce new plants in a sterile growth medium
- Development of more sophisticated plant propagation practices
 - Grafting (segment of tissue is inserted in a cut of another plant)
 - Grows disease resistant plants
- All vegetative propagation techniques contribute to a lack of genetic diversity but produce large numbers of plants with predictable characteristics

SEXUAL TECHNIQUES

- Propagation of plants by selection and germination of seeds results in plant biodiversity and produces large numbers of desirable agricultural plants
- Ripened ovaries and associated structures of flowering plants provide many foods called fruits
- The artificial selection of plants with more desirable seeds and fruit has led to a greater diversity of plant foods
 - Seed banks store seeds from a variety of original plant varieties
 - Factors such as palatability, disease resistance, storage time, size and quantity of produce have been selected to increase agricultural production
- Understanding the structure and function of flowers has led to the ability to control pollination or to cross-pollinate or hybridise varieties of plants to benefit agriculture in a manner that has taken artificial selection one step further

Table 1.7 Economic, social and environmental issues arising from the Green Revolution

Economic issues	Social issues	Environmental issues
in India it transformed the country into a major rice exporter and more than halved the cost of rice	over one billion people are believed to have been saved from starvation	the ratio of crop production to energy use has decreased, with implications for increases in use of fossil fuels and
Mexico became more self-sufficient in agricultural products	Mexican peasant farmers changed attitudes to become more open to the use of scientific ideas in agriculture	the widespread use of fertilisers, herbicides and pesticides, as well as mechanised irrigation, has impacted on biodiversity caused water, soil and air pollution
soy bean production in Brazil and Argentina has dramatically improved as a result of soil treatment; Brazil is becoming a significant exporter of soy beans and, because they are used as stock feed, an exporter of poultry and beef	food security in some countries is at risk because governments are concentrating on export of crops at the expense of crops used to feed the local population	
the use of a new cultivar of rice (IR8), along with the use of fertilisers and pesticides, resulted in the Philippines being able to export rice for the first time in the 20th century	large-scale farming and ownership has meant many poor people have lost control of their land and lives	
corruption has been an impediment to adoption in Africa; Malawi has stood out with its improvements in agricultural production in the 21st century		

- Incidence of polyploidy (cells containing more sets than the diploid set of chromosomes)
- o Conventional breeding along with other changes to agricultural practices result in what has been termed the 'Green Revolution'.
 - o Rice and maize are crops of hybridisation
 - o Maize has both male and female flowers, so it naturally cross-pollinates
 - Removing tassels or stamens from male flowers is a simple way of producing hybrids
 - o Conventional selective breeding used in natural variation is the deliberate attempt to produce variation by exposing plants to mutagens
 - o Insertion of genes from one species into another is known as gene technology or genetic engineering
 - Genetically modified organism is the production of Bt cotton which contains a soil bacterium that is toxic to insect larvae

⇒ 3.2 Impact of scientific knowledge on the manipulation of animal reproduction in agriculture

- o Domestication of livestock relies on mainly sexual reproductive techniques
- o Selective breeding resulted in the development of animals for agricultural purposes
- o Disadvantages:
 - Negative effects on reproductive performance (fertility)
 - Inbreeding due to lack of genetic diversity
- o Artificial insemination is the collection, storage and transport of semen from male animals with desirable characteristics so that more female

can be impregnated and larger numbers of offspring with the desirable characteristics are produced

CELL REPLICATION

INQUIRY QUESTION: How important is it for genetic material to be replicated exactly

1. THE PROCESSES INVOLVED IN CELL REPLICATION

⇒ 1.1 Mitosis

- o The cell replication in which one cell becomes two identical cells
 - They are somatic (body) and allow growth, repair, asexual reproduction and regeneration in multicellular organisms
- o Mitosis and cytokinesis are processes involved in cell replication in eukaryote

Table 2.2 Stages in mitosis and cytokinesis

Stages	Processes occurring
	prophase: the nucleolus disappears and the spindle fibres start to form as the centrosomes move apart; chromatin condenses into chromosomes that already consist of a pair of chromatids joined by a centromere
	late prophase: the chromosomes are fully condensed and the spindle has begun to capture and organise the chromosomes; the nuclear envelope breaks down the spindle also radiates away from the centrosomes towards the cell membrane to form a structure called an aster
	metaphase: chromosomes are all captured by the spindle and brought into alignment along the central axis of the cell (this is sometimes called the equator or metaphase plate though it is not an actual structure) each chromatid pair is attached at the centromere to different spindle fibres so they will move in opposite directions, ensuring that each daughter cell receives a full complement of chromosomes a checking process, called spindle checkpoint, occurs before division can continue
	anaphase: the chromatids separate and move to opposite poles of the cell microtubules not attached to chromosomes extend to lengthen the cell, pushing the poles further apart
	telophase: two new nuclear envelopes form, the chromosomes return to their more diffuse form, the nucleolus reappears and the spindle fibres disappear
	cytokinesis: the final division of cytoplasm occurs in animals this results from actin filaments working with myosin to pinch off the two cells plant cells grow a cell plate that separates the two daughter cells entirely aspects of cytokinesis may commence in late anaphase but complete after telophase

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⇒ 1.2 Meiosis

- o Cell division of one diploid cell to become 4 unique haploid cells that have the potential to become gametes

Table 2.4 Cell division in prokaryotes

Stages	DNA Cell membrane	DNA duplicates	Cell membrane indents	Septum forms
Processes occurring	replication of the circular chromosome and its DNA starts from one point of origin and proceeds in opposite directions it proceeds around the circle until two new loops form; each loop contains one of the original strands of DNA and one new strand	the two circular chromosomes attach to different parts of the cell membrane	the cell elongates (lengthens), separating the two circular chromosomes	a cleavage furrow and septum begin to form across the divide between the two chromosomes

Table 2.3 Stages in meiosis and cytokinesis

Stages	Processes occurring
	prophase: nuclear membrane breaks down; chromosomes condense as pairs of chromatids but also homologous chromosomes (i.e. chromosome pairs) come together adjacent chromatids from the pairs may engage in crossing over, in which exchange of chromosome fragments occurs at structures called chiasmata centrosomes form the spindle
	metaphase: homologous pairs of chromosomes move to the metaphase plate but each member of a pair attaches to opposite spindles; this lining up is random
	anaphase: random segregation of homologous pairs occurs to opposite poles
	telophase and cytokinesis: the nuclear membrane may reform around the two daughter haploid nuclei; chromosomes may diffuse into chromatin and the cytoplasm contents are split by cytokinesis the second part of a meiosis division is similar to mitosis but is occurring in haploid cells; there is no DNA replication in human females this stage of meiosis occurs in the female foetus and is held in suspension until after puberty, when at monthly intervals meiosis in one follicle resumes meiosis only completes after fertilisation (see Chapter 1)
	prophase II: nuclear membrane disappears and chromosomes condense centrosomes move apart and spindle forms and begins to capture chromosomes; sister chromatids are captured by spindle from opposite poles

CELL REPLICATION IN PROKARYOTES

 Table 2.5 Key events in the discovery of the structure and function of DNA

Researcher and date	Discovery
Gregor Mendel (mid-1800s)	discovered the genetic basis of inheritance in pea plants after his work was 'rediscovered' in the early 1900s it triggered the search for the cellular and molecular basis of inheritance
Theodor Boveri (1902) working with sea urchins and Walter Sutton (1903) working with grasshopper testes (independently)	that a full set of chromosomes was necessary for normal development and that Mendel's results can be explained if genes are carried on chromosomes
Thomas Morgan (1910)	discovered sex-linked inheritance such as white-eye colour in fruit flies; this can only be explained if genes are carried on chromosomes
Johann Friedrich Miescher (1869) and his pupil Richard Altmann (who named it nucleic acid in 1889)	Meischer discovered and isolated from nuclei of white blood cells a pure sample of a substance that he called nuclein; later postulated that the substance could be involved in heredity Altmann detected the acidic nature of DNA, changing the name to nucleic acid
Frederick Griffith (1928)	showed that a characteristic (e.g. virulence) could be passed on as a chemical from one bacterium to another (i.e. the bacterium was transformed)
Phoebus Levene (1929)	identified that the basic unit of a DNA macromolecule polymer was a nucleotide consisting of a sugar, phosphate and base identified the four bases : adenine (A), thymine (T), cytosine (C) and guanine (G)
George Beadle and Edward Tatum (1941)	showed experimentally that genes carried 'instructions' to build protein molecules
Oswald Avery (1944)	demonstrated that the bacterial transformation that Griffith discovered could be explained by what happened if DNA was transferred; concluded that DNA is the molecule of inheritance

⇒ 1.3 DNA replication using the Watson and Crick DNA model

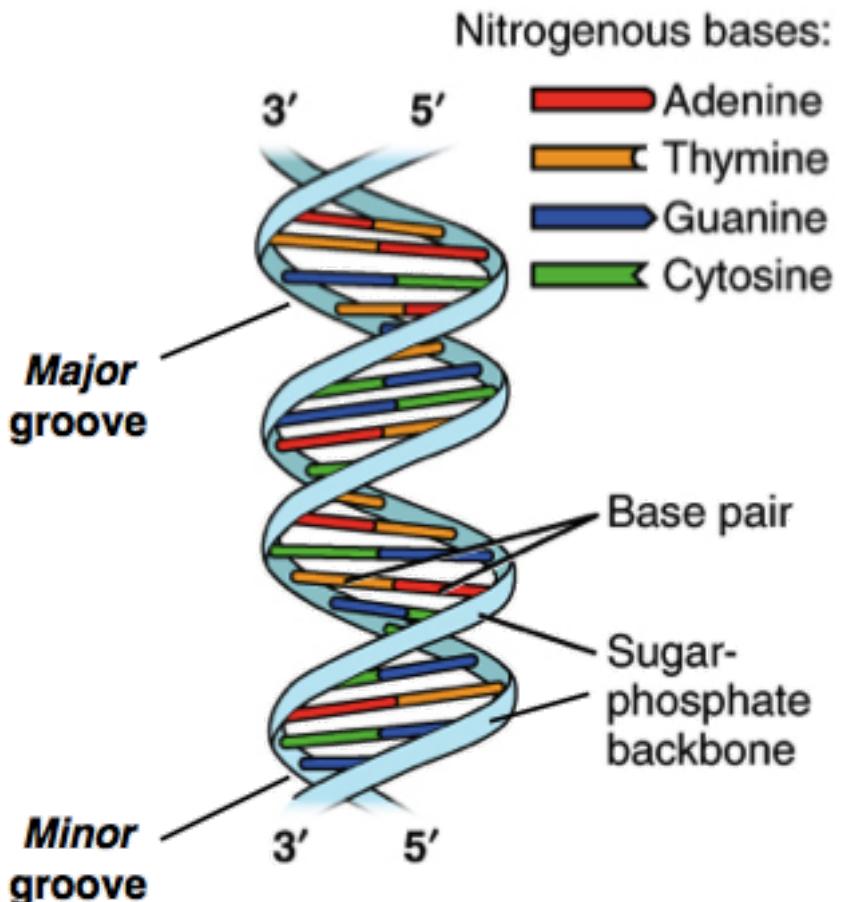
 Here are key events in the lead up to the discovery of the structure and function of DNA

Researcher and date	Discovery
Edwin Chargaff (1940s)	Found that there is always the same amount of adenine as thymine and cytosine as guanine this is called Chargaff's rule
Rosalind Franklin and Maurice Wilkins (1953)	X-ray crystallised forms of DNA to show a helical arrangement
James Watson and Francis Crick (1953)	Built a model of DNA that showed the arrangement of nucleotides which explained ability of DNA to replicate

THE WATSON AND CRICK DNA MODEL

-  X-rays of DNA crystals provided data for model building in the structure of a double helix spiral of two sugar phosphate backbones

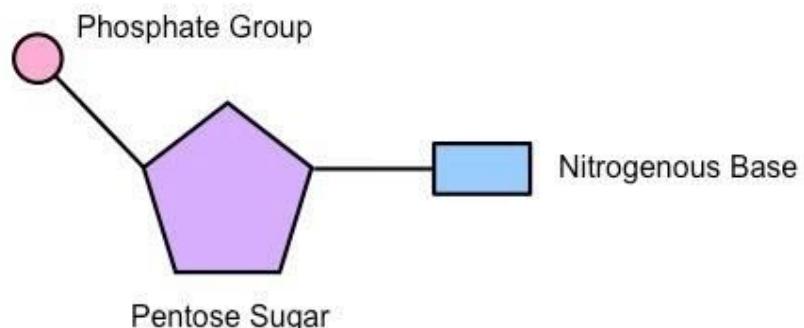
running in opposite directions with internal bases held together in their matching pairs of hydrogen bonds



⇒ 1.4 Nucleotide composition

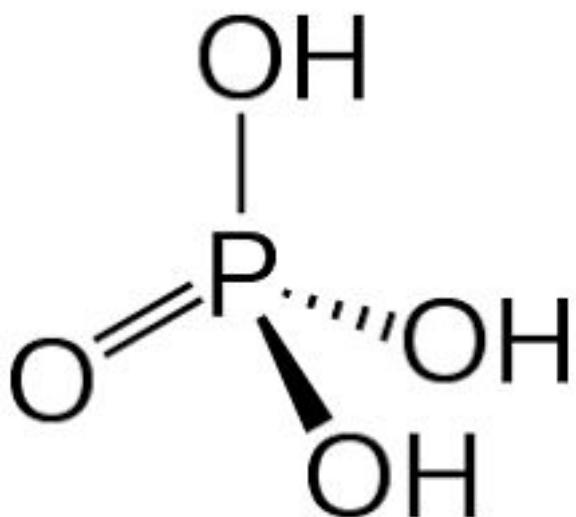
- o NUCLEOTIDES

- Nucleotide monomer in DNA consist of one deoxyribose sugar, one phosphate group and one nitrogenous base



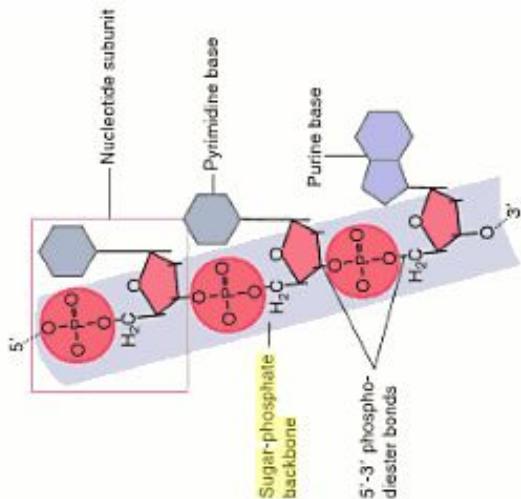
- o PHOSPHATE GROUP

- The phosphate group is derived from phosphoric acid
 - Acids contain hydrogen ions or protons which they release



- o SUGAR-PHOSPHATE BACKBONE

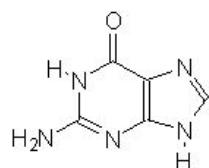
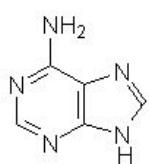
- Alternating sugar and phosphate groups make up sugar-phosphate



- o NITROGENOUS BASES

- The nitrogenous bases that make up DNA exist in two forms: purines and pyrimidines
 - Pyrimidine bases contain four carbon and two nitrogen atoms in a hexagonal ring with other functional groups
 - Purine bases contain two carbon-nitrogen ring bases totalling five carbon atoms and four nitrogen with functional groups attached

The Purines	
Adenine	Guanine
<chem>Nc1nc2[nH]c3[nH]c2[nH]1</chem>	<chem>CN2C=NC3=C2C(=O)NHC3=CN</chem>



The Pyrimidines		
Cytosine	Thymine	Uracil
<chem>Nc1cc[nH]cn1</chem>	<chem>O=C1C=CC(=O)N1</chem>	<chem>O=C1C=CN=CN1</chem>



⇒ 1.5 Pairing and bonding

- Stability of the DNA macromolecule is due to the large number of hydrogen bonds that results from the pairing of A with T and C with G
- The shape of each bases is such that only adenine and thymine align so that hydrogen bonds between hydrogen and nitrogen can form and cytosine and guanine align so that three hydrogen bonds between oxygen and hydrogen and hydrogen can form

⇒ 1.6 The processes involved in cell replication: DNA replication

- DNA replication occurs during the synthesis phase of interphase
- STEPS:
 - 1. DNA double helix uncoils and the strands separate when an enzyme breaks the hydrogen bonds between the matching base pairs
 - 2. The strand terminating in a phosphate group is called the leading strand; the strand that started the uncoiling from its hydroxyl end
 - 3. Units called primers bind with the DNA to start the copying. Primers bind with DNA of the lagging strand at short intervals
 - 4. Nucleotides slot into place in a continuous process along the leading strand. Hydrogen bonding between base pairs helps to ensure faithful replication of the base sequence
 - 5. Enzyme removes all RNA primers and the vacant slots are filled with the appropriate nucleotides. Fragments associated are fused together. They are checked for any errors corrected by enzymes
 - 6. Two identical molecules of DNA have been formed. Each contains one strand from the DNA that acted as a template and one new strand.

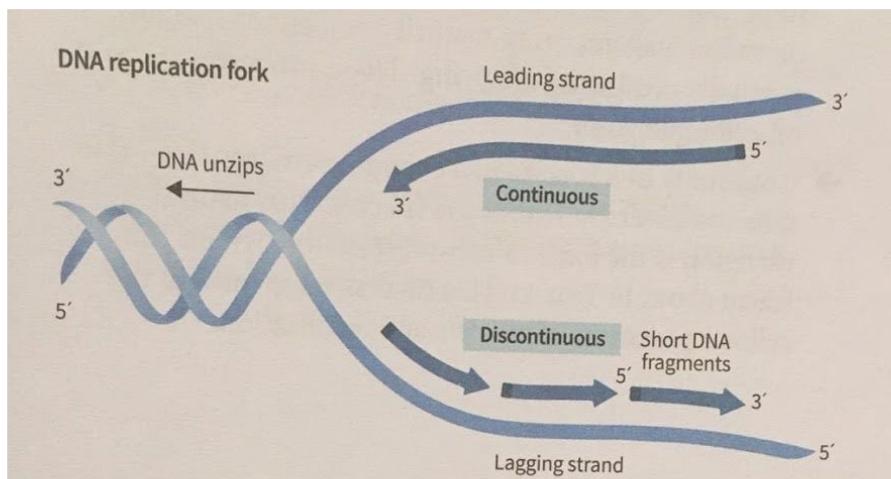


Figure 2.10 DNA replication overview

DNA replication – nucleotides are added to single strands which results in two molecules of DNA from one molecule of DNA. These two double-stranded molecules are the chromatids.

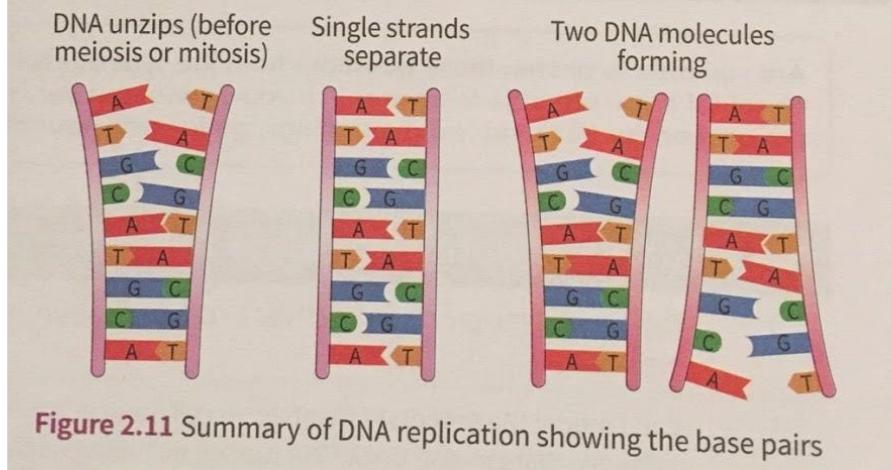


Figure 2.11 Summary of DNA replication showing the base pairs

2. THE EFFECT OF THE CELL REPLICATION PROCESSES ON THE CONTINUITY OF SPECIES

- o Continuity of a species depends on effective reproduction.
 - For most unicellular organisms, reproduction depends on individuals reaching maturity and either sexually or asexually producing offspring
 - These processes depend on cell replication
- o Continuity in a changing environment relies on diversity within a species
 - Diversity depends on the cellular processes of meiosis and fertilisation
- o During an organism's life, cells replicated to provide growth align with repairing and regenerating cells to maintain function

- Processes rely on the integrity of mitosis to ensure faithful copies of cells and their DNA
- Key processes include:
 - DNA replication, including proofing and repairing of mistakes
 - Chromosome duplication to form two chromatids joined by a centromere
 - Chromatids faithfully separating. Into different daughter cells
 - Cytokinesis ensuring both daughter cells are viable by containing sufficient cytoplasm to function
- o Sexual reproduction relies on the transmission of genes
 - Meiosis is the key cell divisions essential for the production of gametes
 - Processes include
 - Crossing over of paired chromatids, resulting in exchange of genes and DNA and new combinations of genes aligning a chromosome
 - Random and independent segregation of chromosomes
 - Halving of the number of chromosomes

DNA AND POLYPEPTIDE SYNTHESIS

INQUIRY QUESTION: Why is polypeptide synthesis important?

1. REPRESENTATIONS TO MODEL AND COMPARE THE FORMS IN WHICH DNA EXISTS IN EUKARYOTES AND PROKARYOTES

⇒ 1.1 Modelling forms of DNA

- o Modelling often involves simplification so that the key processes are understood and not overwhelming

PROKARYOTE	EUKARYOTE
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Most genetical material is in a supercoiled DNA found in the nucleoid Protein help to form some loops or bends	DNA found in the nucleus as chromatin Protein package is a histone
DNA replication starts from a single origin progresses in both directions	Replication occurs in interphase before mitosis
12% is non-coding DNA	98% is non-coding DNA
Has one copy of each gene	Homologous pairs contain two copies of each gene
DNA replication is fast	Relatively slow
No nucleus	DNA found in membrane-bound nucleus
No organelles	Organelles are membrane-bound

- o Similarity is that they both have ribosomes

2. MODELLING THE PROCESS OF POLYPEPTIDE SYNTHESIS

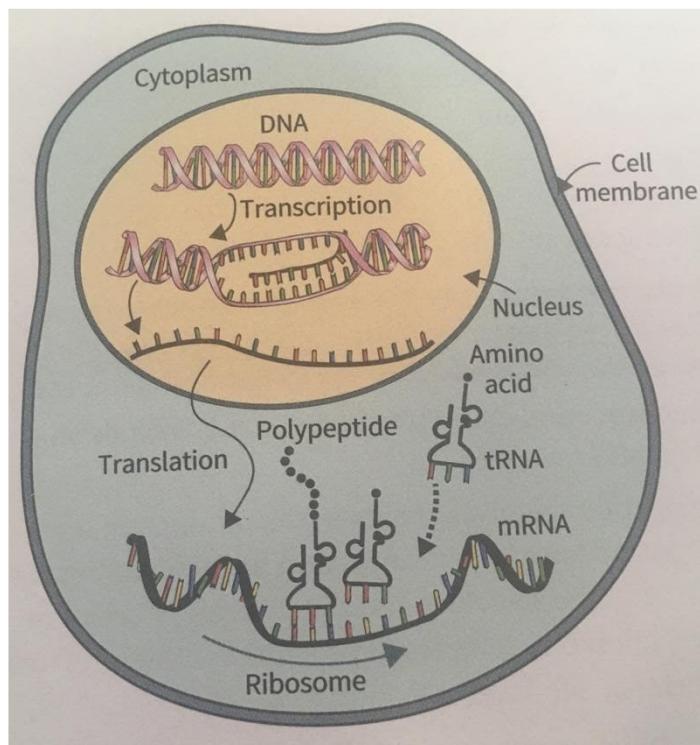
⇒ 2.1 Polypeptide synthesis

- o GENES
 - Genes carry the inherited information give organisms their characteristics and enable them to function
 - Major part of gene expression is polypeptide synthesis
- o POLYPEPTIDES
 - Polypeptide is a long chain of amino acids that make up proteins
 - Peptide bonds join amino acids into the chain
- o RIBONUCLEIC ACID (RNA)
 - RNA is nucleic acid made up of nucleotides
 - Sugar in RNA is ribose

FORM OF RNA	DESCRIPTION AND ROLE
Messenger RNA (mRNA)	Single strand of RNA that complements the base sequence of the DNA which is the template triplets of bases are codons that link to a specific for amino acid It is bound to proteins It transfers the genetic code from DNA in the nucleus to the ribosome in the cytoplasm

Transfer RNA (tRNA)	Convoluted and relatively short segment of RNA contains a bond to an amino acid and, at the other end, an anticodon triplet of bases slots into mRNA at a ribosome 'reads' the genetic code to insert amino acids into the polypeptide chain
Ribosomal RNA (rRNA)	Forms a complex with protein to make. Up the large and small ribosomal subunits that come together during translation and then disband Holds mRNA in place for insertion of tRNA

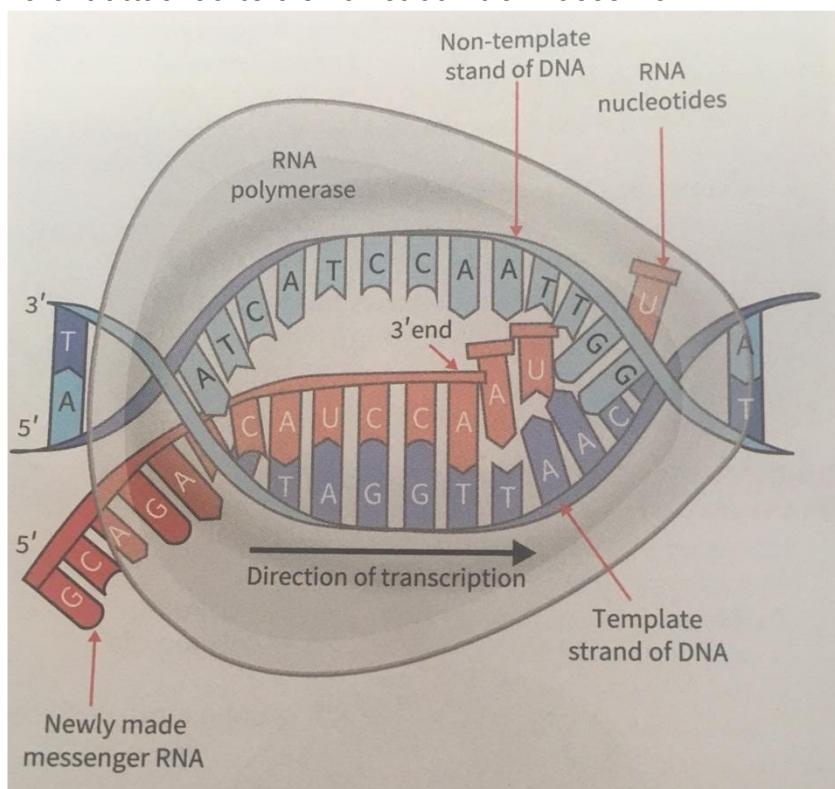
⇒ *2.2 Transcription*



- o DNA is transcribed and then translated into a

- polypeptide chain
- o Both steps involve RNA that carry a sequence of triplets of nitrogenous bases
- o Enzymes facilitate the process and ensure the accuracy of the conversion of genetic code, as stored on the DNA, to functional polypeptides and hence proteins
- o First process is transcription of DNA into messenger RNA (mRNA)
- o The transcription process is how the genetic code stored in DNA is copied into another form, mRNA

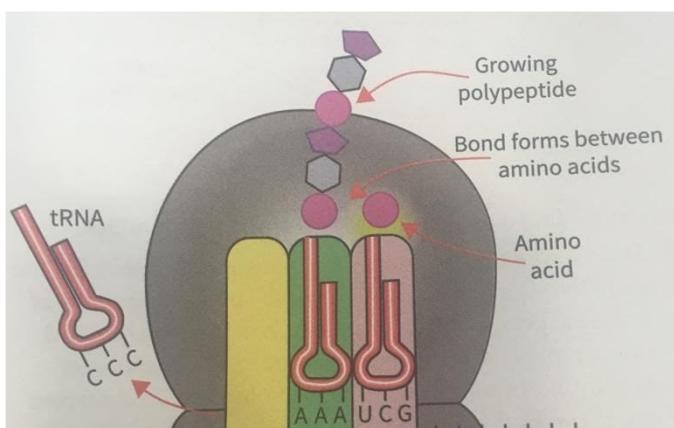
- Transcription is a rapid process that occurs in quick succession when polypeptides are needed
- DNA in the nucleus carries the genetic code.
- DNA is unzipped and then a starter is identified and one strand acts as the template for the manufacture of one strand of mRNA. This is processed and carried out through the nuclear pore into the cytoplasm
- Enzymes such as RNA polymerase assist the manufacture of mRNA by unwinding the DNA, moving the RNA nucleotides into position and promoting attachment and elongation
- Each triplet of bases is called a **codon**
- Start and stop codons control the part of the DNA template that is 'read' in the process of transcription
- mRNA leaves the nucleus through the pores and enters the cytoplasm. There it attached to a small subunit of ribosome



⇒ 2.3

Translation

- The process occurs on the ribosomes
- Uses mRNA codons as the template for coding for a sequence of amino acids in a polypeptide chain
- tRNA anticodons carry the appropriate amino acid into place
- translation is often divided into three stages: initiation, elongation and termination



- o INITIATION
 - Once in the cytoplasm, a 'start' codon on the mRNA binds. Between the small and the large ribosome subunits
 - Start codon on the first part of the mRNA contains the AUG bases. This matches with the tRNA anticodon UAC and carries the amino acid methionine
- o ELONGATION
 - Ribosome acts like a conveyer belt, moving the mRNA along, and the large. Subunit has grooves into which fit two tRNA units that each carry an amino acid
- o TERMINATION
 - Terminal sequence of the mRNA stops the elongation process and releases polypeptide chain

⇒ *2.4 Importance of mRNA and tRNA in transcription and translation*

- o In polypeptide synthesis DNA remains within the nucleus. The ribosomes are in the cytoplasm. mRNA is essential to carry the genetic code stored in DNA from the nucleus to the ribosomes
- o Three nucleotides in mRNA form a codon that match with an anticodon
- o AUG is the start codon
- o Stop codons include UAG, UAA and UGA which terminate the polypeptide chain
- o tRNA carries a specific amino acid to be joined into the polypeptide chain
- o Correct matching of the codon on the mRNA to the anticodon on the tRNA is critical for the correct sequence of amino acids in the polypeptide chain

⇒ *2.5 Analysing the function and importance of polypeptide synthesis*

- o Inheritance and species continuity depend on polypeptide synthesis
- o Polypeptide synthesis provides the means for the inherited qualities that are carried in the DNA to be mobilised into cellular structure and function

⇒ 2.6 Assessing how genes and environmental affect phenotypic expression

- o Total base composition and arrangement of the DNA in an organism is called its genome
- o Genetic composition is called the genotype
- o The traits of an organism are qualities such as height and eye colour which is known as the phenotypes

Researcher(s)	Discovery
Associate Professor Jeff Craig	Environment in the womb has a large impact on later health
Professor Brian Byrne	Investigated reading ability and literacy development and found impact of teacher not as high as expected as twins in separate classrooms developed similar literacy skills as those in the same classroom
Professors Sam Berkovic	Dispelled belief that most forms of epilepsy were due to injury and demonstrated that one serious form was not a result of immunisation but was caused by a gene mutation a diet avoiding glucose was demonstrated to be an effective treatment in some cases
Perminder Sachdev and Julian Trollor	OATS is investigating environmental versus genetic factors that contribute to amyloid plaque development

3. THE STRUCTURE AND FUNCTION OF PROTEINS IN LIVING THINGS

- o The sequence of amino acids on the polypeptide chain(s) determines the three-dimensional shape of a protein
- o Proteins are essential to the structure, function and regulation of the body's cells, tissues and organs
- o Protein structure is described using four levels: primary, secondary, tertiary and quaternary
- o Proteins spontaneously fold into three dimensional shapes that is determined by amino acid sequences
- o Proteins contain functional groups in the amino acids with varying chemical properties

Function
Catalysts (enzymes) <ul style="list-style-type: none"> - Promote and control metabolism

- Control the chemical reactions

Transport

- Allows the selective entry and exit of materials
- Proteins may be involved in active or passive transport
- Some proteins facilitate in the transport of body fluids

Storage proteins

- Some contain molecules that hold metals that help store oxygen
- Other proteins store amino acids for the growth and development of embryos, foetuses and infants

Mechanical support

- Proteins provide the strength and varying levels of elasticity needed in many tissues and organs
- Many of these proteins are extracellular

Immune protection

- Detection and destruction of foreign microbes is facilitating by the structure and function of antibody proteins

Generate movement

- Cells can change shape because of proteins that interact to use energy to produce force and movement

Messengers (hormones)

- Control of growth and differentiation

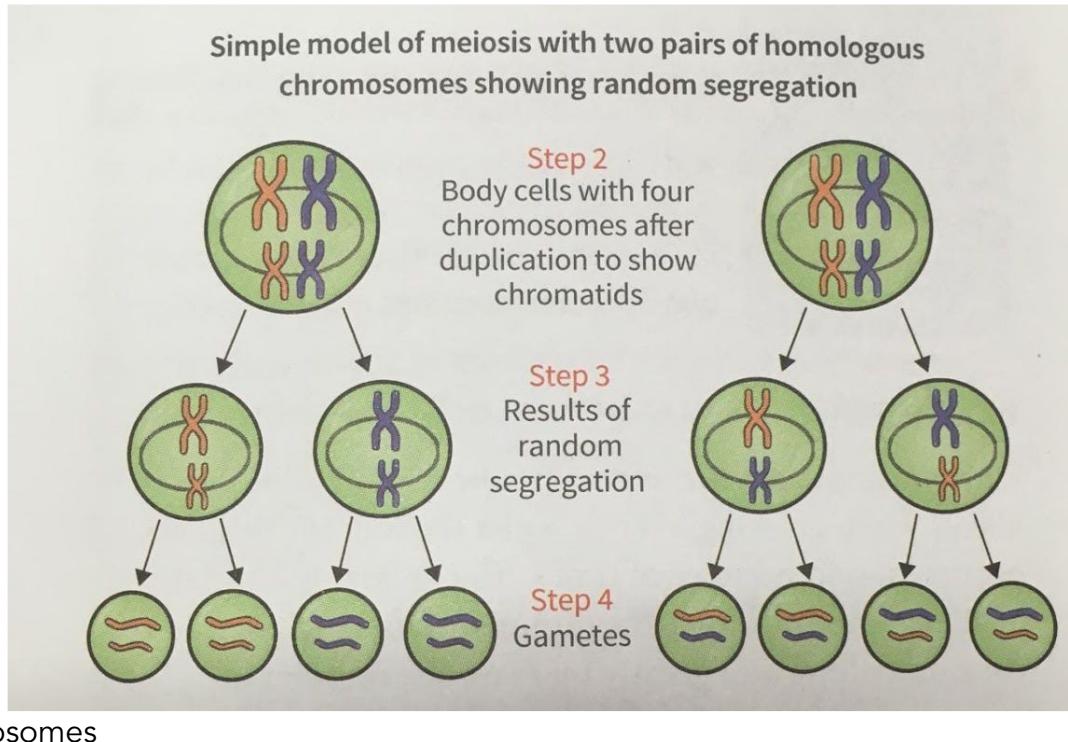
GENETIC VARIATION

INQUIRY QUESTION: How can the genetic similarities and difference within and between species be compared?

1. VARIATIONS IN GENOTYPES OF OFFSPRING

⇒ 1.1 Predicting variations in the genotypes of offspring by modelling meiosis

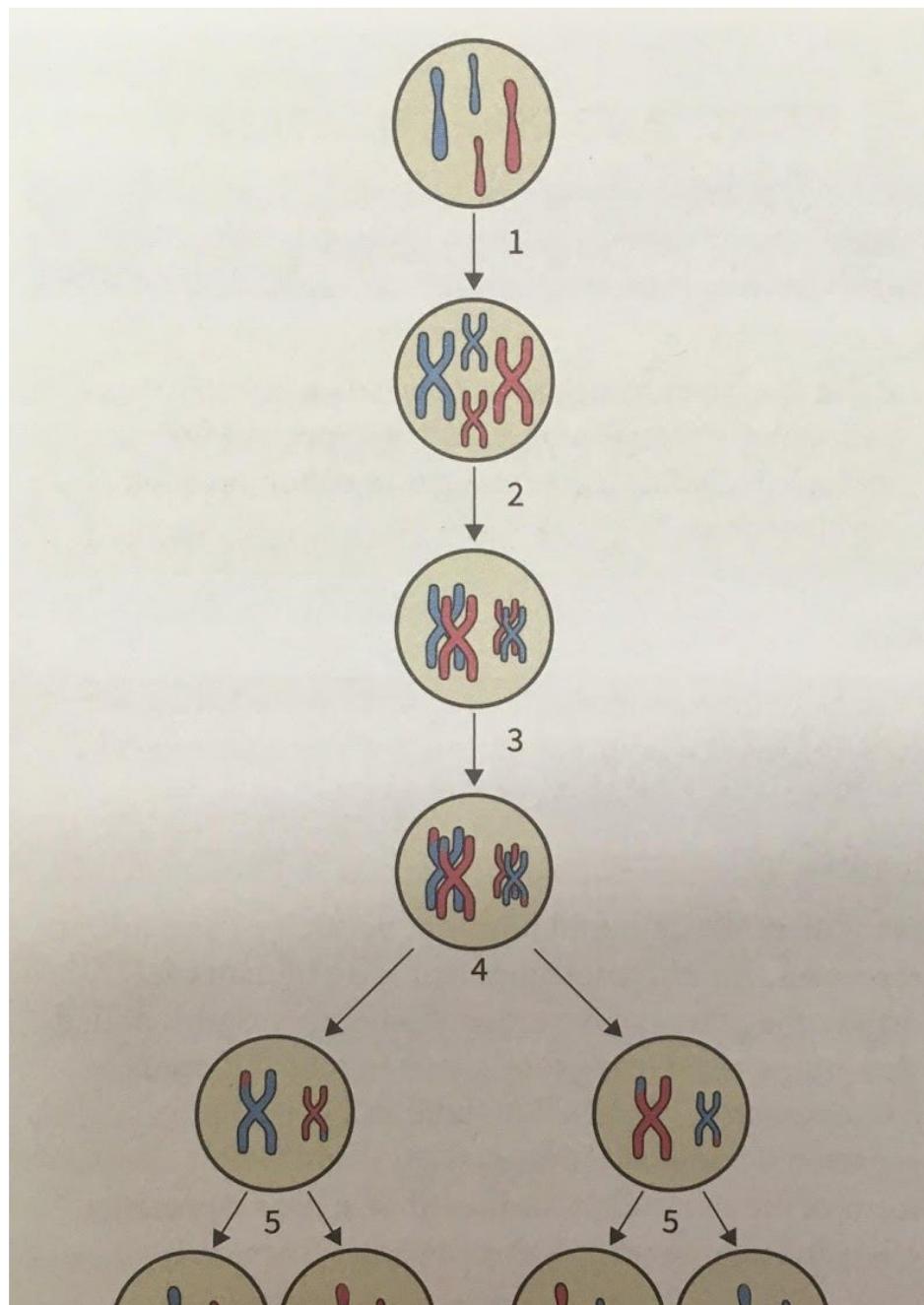
- o Sexual reproduction depends on fertilisation which will also be a source of variation which can be mathematically predicted
- o Diploid organisms possess homologous pairs of chromosomes. The autosomes are the chromosomes that carry genes for normal development and appear as identical pairs in a karyotype
- o Humans have 23 homologous pairs, that is 22 autosomal pairs and one pair of sex



⇒ 1.2 Random segregation

⇒ 1.3 Crossing over of homologous chromosomes

- o Crossing over involves chromatids breaking at the same point and re-joining in two reciprocal non-parental combinations
- o Combined with random segregation of homologous chromosomes, many possible combinations of alleles located along the same chromosome can be predicted.



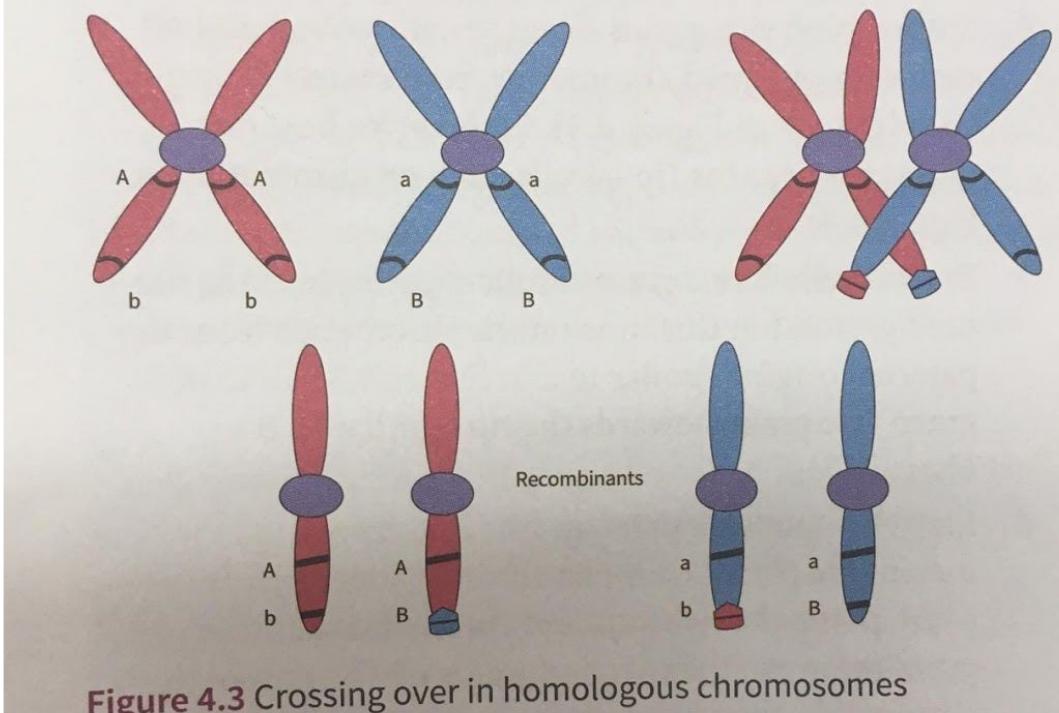


Figure 4.3 Crossing over in homologous chromosomes

⇒ 1.4 Fertilisation

- o Predicting variations of genotypes from fertilisation is demonstrated by analysing a Punnett square diagram
- o The intersection of rows and columns contains the possible genotype in male and female offspring

	X	Y
X	XX	XY
X	XX	XY

⇒ 1.5 Mutations

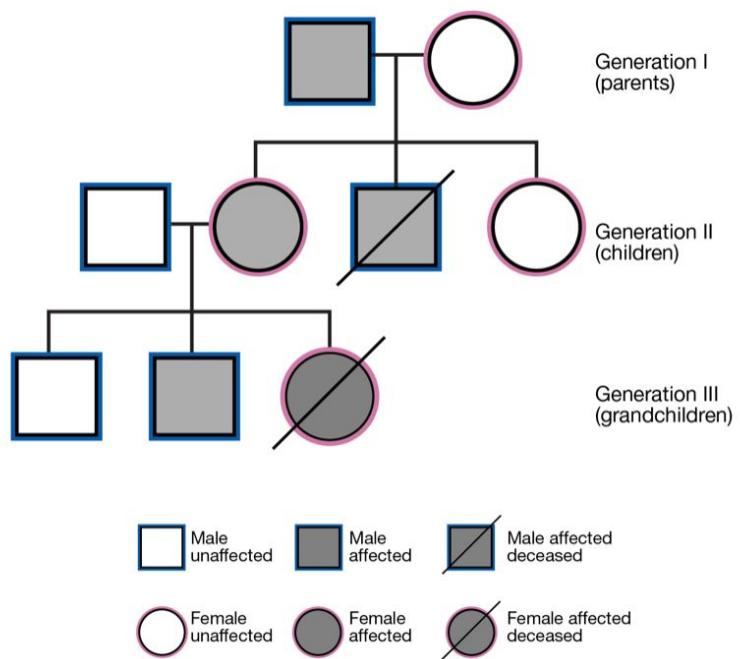
- o Caused by genotype variation
- o They result in new alleles
- o There is a change in the DNA
- o Mutations can be both harmful and beneficial to an organism
- o Genotypes for a trait in an organism are expressed as letters that represent the alleles
- o The letters are the same then it is said to be homozygous
- o If the letters are different then it is heterozygous

2. FORMATION OF NEW COMBINATIONS OF GENOTYPES PRODUCED DURING MEIOSIS

⇒ 2.1 Autosomal inheritance

- o Inheritance pattern occurring similarly in males and females because alleles responsible are carried on autosomes

- Breakthrough was by Gregor Mendel's pea plants
- It was apparent that for each phenotype, one form of allele was dominated
- One trait will be dominant whilst the other is recessive
- These are called Mendelian ratios
- Pedigree diagrams are another method of showing inheritance of characteristics over at least one generation



- Circles represent females, squares represent males, shading indicates those expressing the trait, horizontal lines show parents/siblings

⇒ 2.2

Table 4.5 Sex-linked inheritance in fruit flies

Male gametes →
Female gametes ↓

X^r

X^R

Y

X^r

$X^r X^R$

$X^r Y$

X^r

$X^r X^R$

$X^r Y$

Key to table

$X^r X^r$: white-eyed female

$X^R Y$: red-eyed male

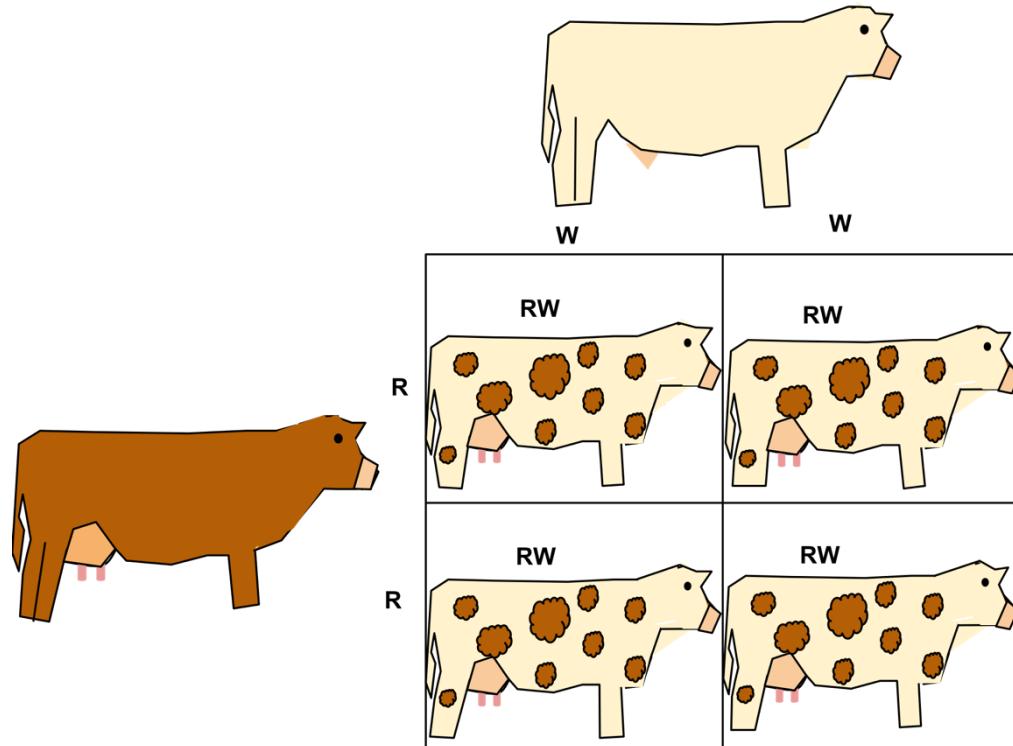
Results of the cross: all red-eyed heterozygous (carrier) females and all white-eyed males

Sex-linked inheritance

- Some genes are carried on the sex chromosomes
- This inheritance pattern is called sex-linked and is associated with gender
- Male Y do not carry alleles that are carried on X

⇒ 2.3 Co-dominance

- When both alleles are expressed, they are described as co-dominant



⇒ 2.4 Incomplete dominance

- The heterozygous form results in a blend of both allele characteristics

Incomplete Dominance- F₁ generation

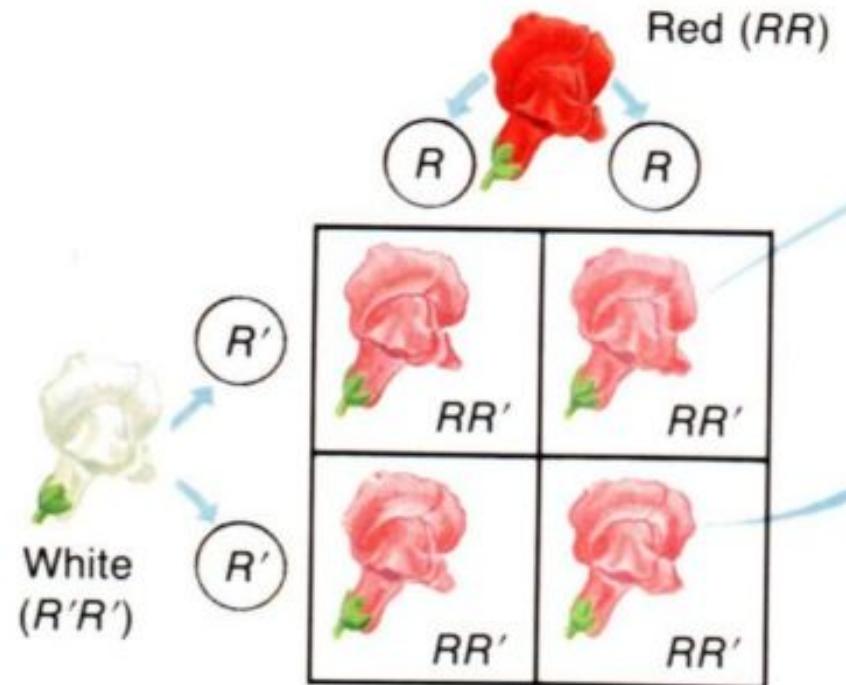


Fig.3(a). Incomplete Dominance

⇒ 2.5 Multiple Alleles

- Some traits have more than two forms within the gene pool

Blood Type	Genotype			
A	I ^A	I ^A	or	I ^A I ^O
B	I ^B	I ^B	or	I ^B I ^O
AB	I ^A	I ^B		
O	I ^O	I ^O		

3. REPRESENTING FREQUENCIES OF CHARACTERISTICS IN A POPULATION

⇒ 3.1 Examining frequency data

- o Genetic diversity between species is based on comparison of overall genomes
- o Gene pool is all the copies of all the alleles of all the genes in a population
 - It is a stock of different genes in an interbreeding population
 - A large gene pool is an indicator of genetic diversity within a species
- o Allele frequencies is the proportion or percentage of all possible alleles at a locus of one particular allele
 - It indicates the genetic diversity or richness of a gene pool
 - The allele frequency is expressed as a proportion or percentage of the allele/gene frequency in a population or species
- o Genotypes are used to provide data for calculation of allele frequency
- o Phenotypes are used to collect data on allele frequency
- o It is possible for some alleles in a population to have no variation
 - Occurs when a particular characteristic is essential for survival
- o Factors such as genetic isolation, migration, mutation, natural and artificial selection and chance can affect allele frequencies
- o Population genetics is involved with the study of allele frequencies in a population

⇒ 3.2 Analysing single nucleotide polymorphism (SNP)

- o SNPs are variations in one nucleotide base, such as thymine being substituted by cytosine on a particular point in the DNA
- o SNPs may occur in coding and non-coding sections of the genomes but are most common in the sections between genes
- o SNPs account for 90% of naturally occurring human DNA variation and occur on average once every 300 nucleotides
- o When SNPs occur, they may impact a gene's function and directly cause a disease
- o Alternatively, they can be used as biological markers to help scientists locate genes associated with disease
- o SNPs can also be used in sustainable agriculture

⇒ 3.3 Identifying trends, patterns and relationships

- o Trends are identified by comparing data over time or across different locations or population groups
- o Patterns are detected when there are similar trends such as an increase in allele frequency over time for several alleles
- o Relationships occur when a trend can be linked to external factors. Changes in human skin pigmentation have been linked to adaptations for protection from skin cancer and the ability to make vitamin D from exposure to UV radiation

⇒ 3.4 Limitations in data

- Collecting data on characteristic frequency is dependent on observational data, obtaining accurate family records, and decisions about aspects of particular phenotypes
- Collecting data on SNPs provides an accurate snapshot for specific alleles of genes but the data collection relies on sampling

INHERITANCE PATTERNS IN A POPULATION

INQUIRY QUESTION: Can population genetic patterns be predicted with any accuracy?

1. USE OF TECHNOLOGIES TO DETERMINE INHERITANCE PATTERNS IN A POPULATION

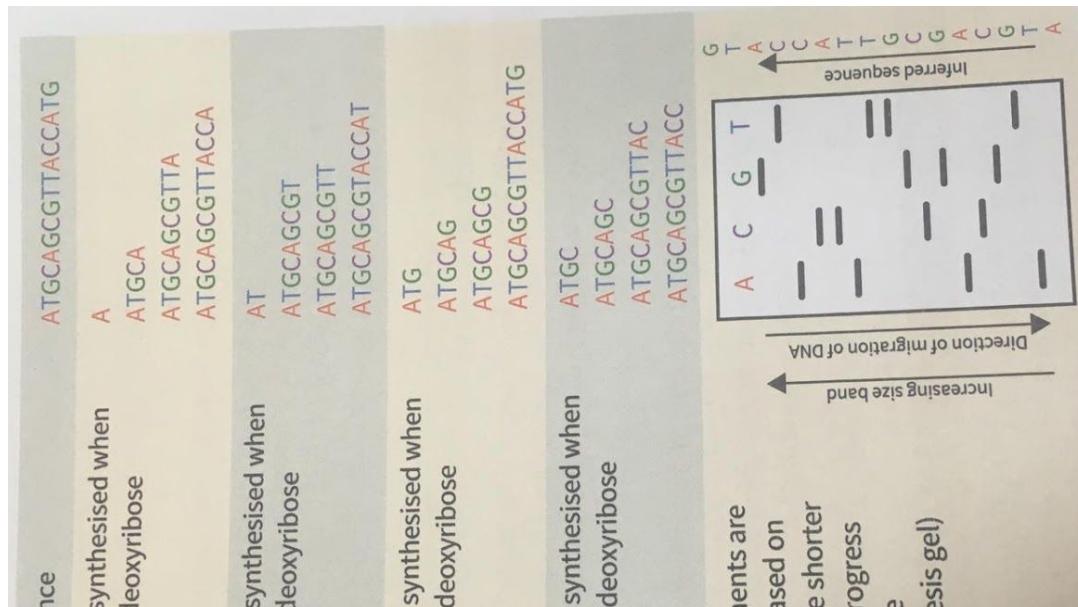
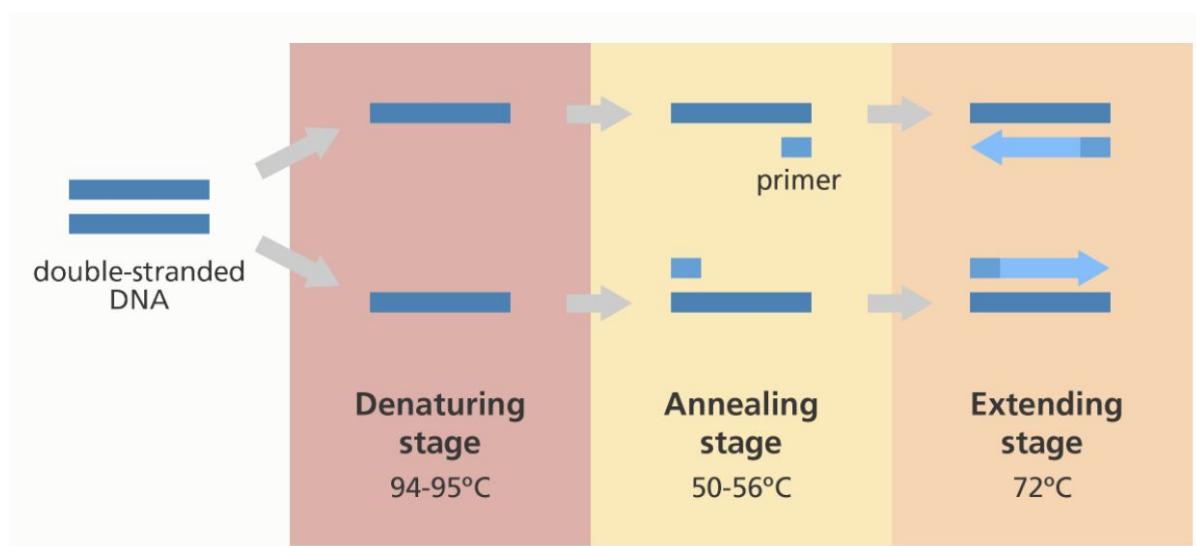
⇒ 1.1 DNA sequencing

- DNA sequencing is a complex set of technology dependent processes used to determine the sequence of nucleotide bases in a genome
- It is the process used to determine the order of nucleotides in DNA
- Involves the following processes
 - DNA is denatured into separate strands
 - Primers and specific enzymes are used to make copies of the labelled strands
 - This amplifies DNA through polymerase chain reaction (PCR)
 - PCR is a form of artificial DNA replication that uses specific enzymes and heating and cooling to make unlimited copies of a specific DNA sequence
 - DNA strands are broken into labelled pieces by using particular enzymes called nucleases in a process known as hydrolysis
 - Pieces of DNA are separated so the labelled bases can be identified in order: one traditional method of separation of the fragments is electrophoresis
- From this process, it is possible to discover genetic patterns and changes in populations, such as mutations, SNPs and other changes in DNA nucleotide sequences

1. Denaturing – when the double-stranded template DNA is heated to separate it into two single strands.

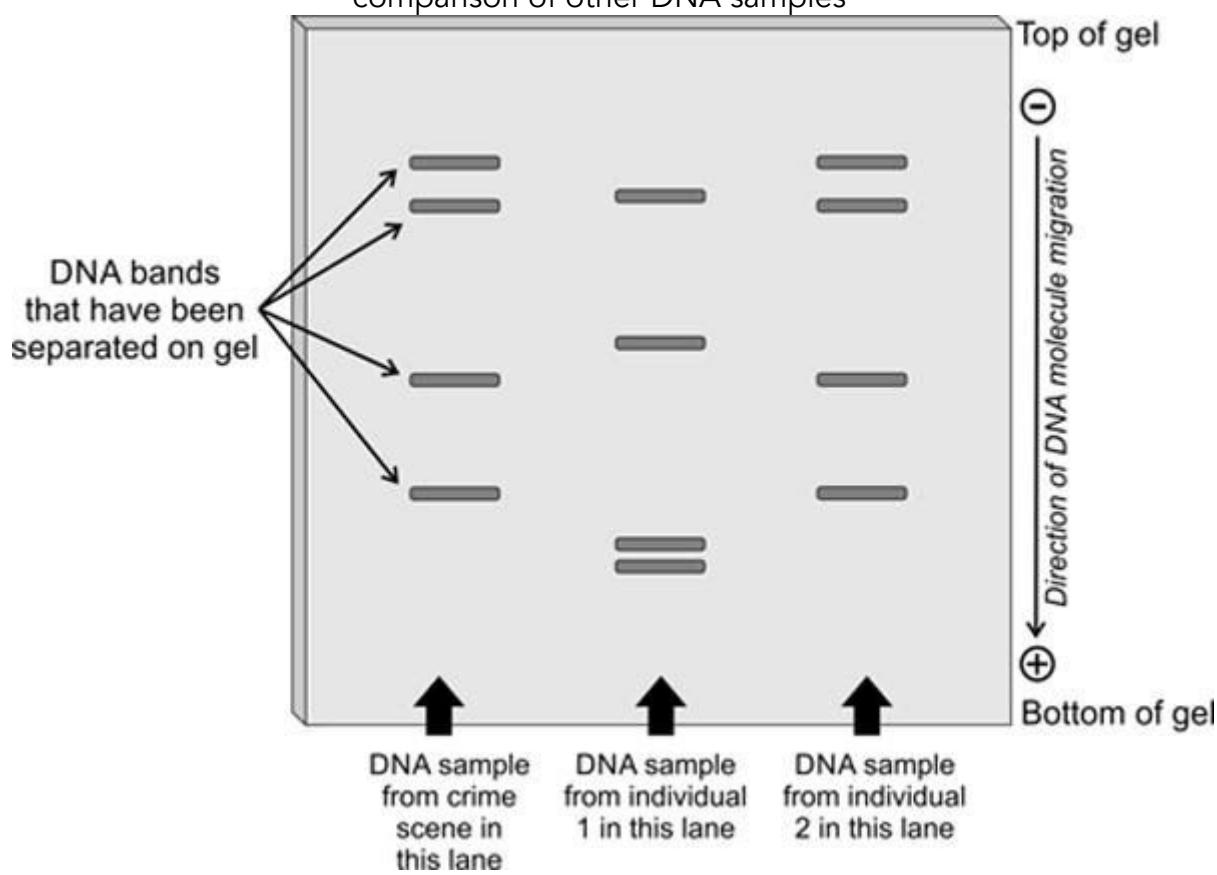
2. Annealing – when the temperature is lowered to enable the DNA primers to attach to the template DNA.

3. Extending – when the temperature is raised and the new strand of DNA is made by the Taq polymerase enzyme.



⇒ 1.2 DNA profiling

- Also known as DNA fingerprinting
- It is the process used to determine an individual's DNA characteristics, often for the purposes of paternity testing, genealogy, identification of remains or criminal investigation
- Used in forensics and in the identification of family relationships
- Criminal investigation relies on the purity of the DNA samples
- The process involves the following steps:
 - Collection of biological samples, DNA extraction and purification
 - Restriction fragment length polymorphism (RFLP) where lengths of DNA have been cut by restriction enzymes in specific locations into fragments that are separated and compared. The fragments vary in length between individuals as they contain variable number tandem repeats
 - Fragments are separated through electrolysis
 - Fluorescent dyes may replace radioactive probes
 - They are transferred to a membrane in a process called blotting
 - X-ray film is used to detect a radioactive pattern followed by a comparison of other DNA samples



2. USE OF DATA ANALYSIS FROM A LARGE-SCLAE COLLABORATIVE PROJECT

⇒ 2.1 Population genetics in conservation management

- The study of the frequencies of variants of alleles, the process bringing about changes in alleles, population structures and divisions
- It is the study of genetic characteristics of populations and how and why the allele frequencies change over time and space
- Many genes in a population are polymorphic meaning there exists more than one
- Data analysis of information contributes to decisions about conservation of endangered species, provides evidence for the genetic component cause of some diseases and provides evidence of human evolution and migration
- Diversity in a gene pool may result from population size, mutation, genetic drift, natural selection, diverse environments and population movements resulting in gene flow
- TASMANIAN DEVIL
 - Population of Tasmanian Devil is under threat from the Devil Facial Tumour Disease which is contagious
 - An unstable climate 5000 years ago caused a dramatic reduction in population, distribution and diversity
 - Lack of genetic diversity (only 1 million SNPs) is a factor in susceptibility to the disease
 - Devils have low diversity in the genes
 - Genetic studies have allowed researchers to observe mutations found in cancer cells
 - Genetic investigations are focusing on behavioural adaptations to reduce the spread of cancer
 - It allows cancer-free preserves of the population to be established

⇒ 2.2 Inheritance of a disease or disorder

- Almost all non-infectious disease has a genetic component
- Genetically isolated populations such as Iceland and Finland have proved useful in the identification of rare recessive disease genes

Description of the project	Trends, patterns and relationships	Strategies, goals, outcomes, data analysis
WTCCC Large series of case-control studies combines with research Map variation in human genotypes across the UK	Compared DNA of people with tuberculosis, coronary heart disease, type 1 diabetes, type 2 diabetes, rheumatoid arthritis, Crohn's disease, bipolar disorder and	Compared 500000 SNPs of people with single disease findings included a clear genetic link to obesity and three new genes linked to type 2 diabetes

	hypertension with 3000 healthy controls	Major gene region on chromosome 9 was verified Four chromosome regions containing genes that can predispose people to type 1 diabetes were discovered
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⇒ 2.3 Human evolution

- o Modern humans share a recent link with the Neanderthals and the Denisovans
- o Analysing data shows us the similarities and differences in organism alleles from different populations

GENETIC CHANGE

MUTATION

INQUIRY QUESTION: How does mutation introduce new alleles into a population?

1. HOW A RANGE OF MUTAGENS OPERATE

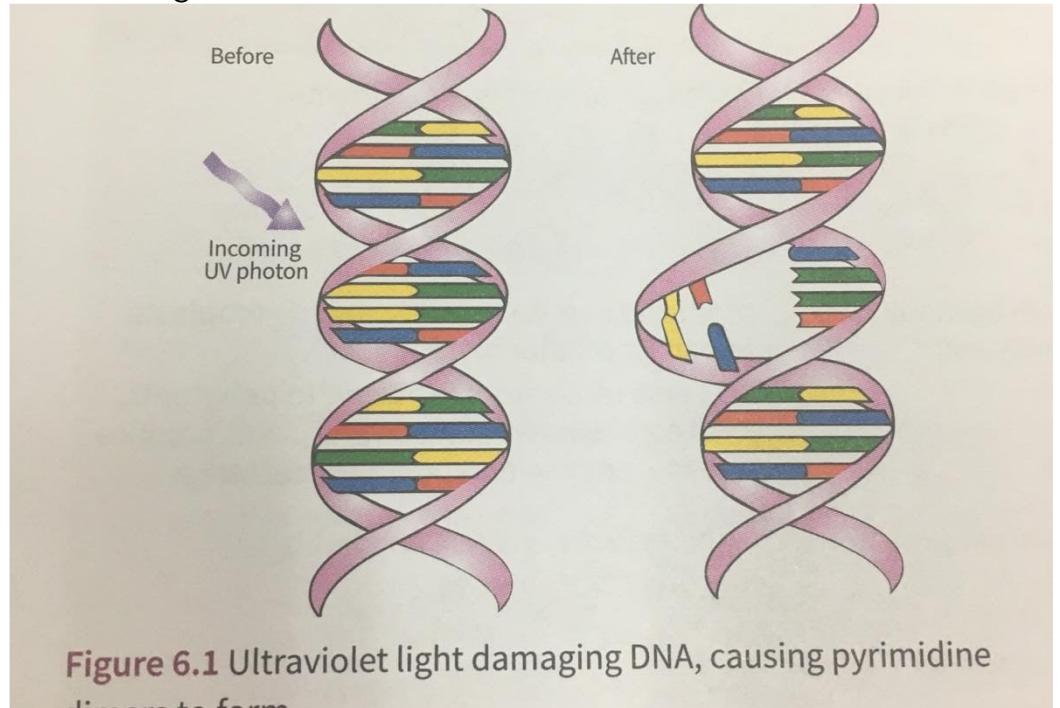
⇒ 1.1 Mutations

- o Mutations are permanent changes in the DNA or RNA of a cell
- o It includes changes to the nucleotide sequence of DNA or RNA
- o DNA replication and synthesis have built-in checks and repair pathways
- o A mutation is permanent when the repair process fails to correct the error
- o Mutations may harm cells and cause diseases like cancer
- o Mutations may arise spontaneously
- o Mutagens are chemicals or energy forms such as radiation that promote changes in the genetic material (DNA) and hence mutations
- o Mutagens are often classed as physical (electromagnetic radiation) chemical (cigarette smoke) or natural (viruses)
- o Mutations can be harmful, neutral or positive
- o Endogenous mutagens come from the inside the organism's body

- Exogenous mutagens come from outside the organisms body

⇒ 1.2 Electromagnetic radiation sources

- The oscillating electric and magnetic fields, which travel at right angles to each other in a bundle of energy called a photon
- It refers to a range of photons carrying energy in the form of electromagnetic fields through time and space
- Many high radiation energy types (small wave length) cause mutations such as UV, X-rays and Gamma rays
- UV LIGHT
 - Damages living tissues
 - Makes skin cells cancerous
- X-RAYS
 - High frequency causes damage to cell DNA causing cell death or uncontrollable division which causes cancer
- GAMMA RAYS
 - An ionising radiation



- Break down DNA molecules, damages base and causes large chromosomal deletions
- Increases potential of cancer to develop

⇒ 1.3 Chemicals

- Substances that increase the likelihood of mutations
- Cause changes to the DNA nucleotide bases

Table 6.1 Chemical mutagens

Chemical mutagen	What it does	Possible source
5-bromo-deoxyuridine	mimics nitrogenous bases	exposure results from it being used to detect actively dividing cancer cells in living organisms
nitrogen mustards (from World War I and II), nitrosoureas, alkyl sulfonates, triazines and ethylenimines	alkylating agents produce mutations by adding small hydrocarbon groups, such as methyl groups, to either the bases or the backbone phosphate groups of DNA	exposure to mustard gas used as a chemical weapon or a component (and side effect) of chemotherapy
deaminating agents (e.g. dimethyl nitrosamine)	removes amine groups from bases, such as converting cytosine bases to uracil, producing mutations	produced in an animal's stomach after eating foods containing nitrous acid or nitrite
intercalating agents such as acridine orange, proflavin and ethidium bromide	insert themselves into DNA, 'stretching' it and causing frameshift mutations	used in laboratories as dyes or mutagens
DNA adducts such as polycyclic aromatic hydrocarbon (PAH)	DNA adducts are a form of DNA damage in which the chemical mutagen attaches to the DNA	found in coal and tar deposits and released by the incomplete combustion of organic matter

⇒ 1.4 Naturally occurring mutagens

- Physically or chemically promote mutations
- Can come from biological and non-living origins
- Microbes, plants, animals, chemicals
- HPV and HIV can change the functioning of cells and trigger cancers
- HPV infectious disrupt the normal functioning of cells
- HIV and HCV affect the body's immune system

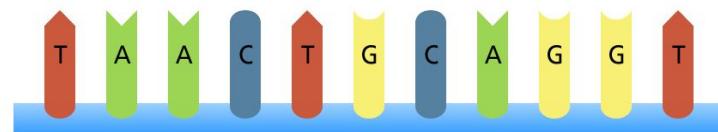
2. THE CAUSES, PROCESSES AND EFFECTS OF DIFFERENT TYPES OF MUTATION

⇒ 2.1 Point mutation

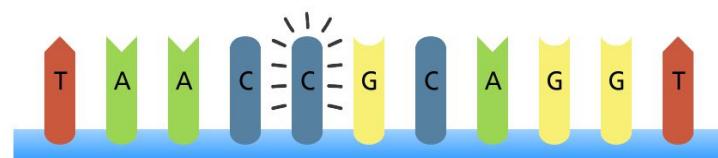
- A change in one nucleotide in a DNA sequence is called a point mutation
- It is when one base is substituted for another
- If a substitution of a nitrogenous base occurs in a coding part of DNA it could result in the substitution of an amino acid for another polypeptide synthesis
- Substitution in a non-coding region could alter the expression of a gene
- Other changes from point mutations include, insertions or deletions which may result in frameshift mutations
- Point mutations usually occur spontaneously during DNA replication when a cell is undergoing mitosis or meiosis

No mutation	Point mutations			conservative	non-conservative
	Silent	Nonsense	Missense		
DNA level	TTC	TTT	ATC	TCC	TGC
mRNA level	AAG	AAA	UAG	AGG	ACG
protein level	Lys	Lys	STOP	Arg	Thr
					basic polar

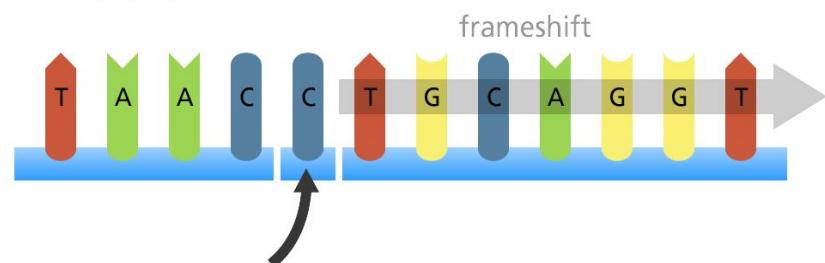
Original sequence



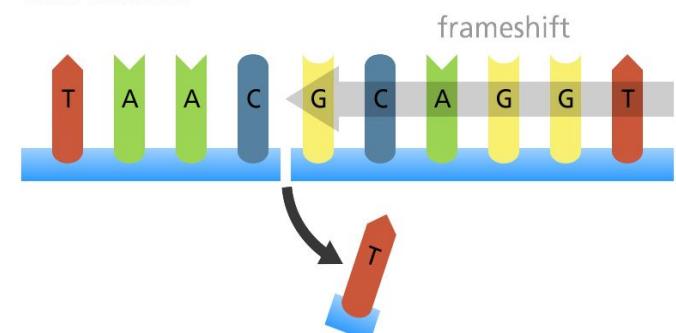
Base substitution



Base addition



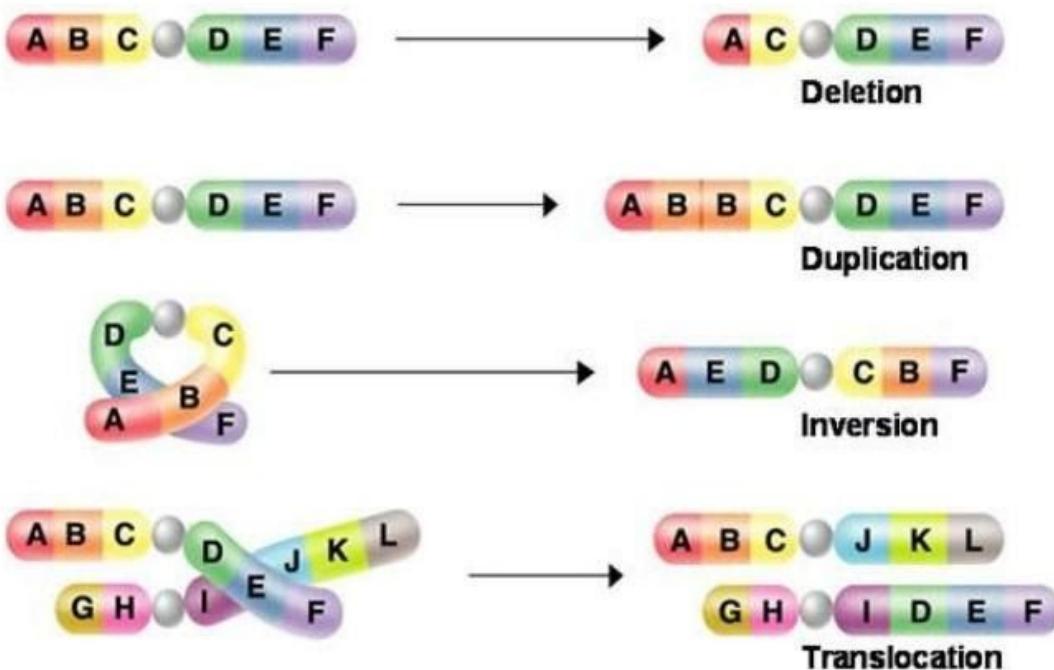
Base deletion



⇒ 2.2 Chromosomal mutations

- Occurs when a cell is undergoing cell division
- Occur due to exposure to radiation or chemicals
- Occur by chromosome rearrangement and changes in chromosome numbers
- Main types include
 - Deletion
 - Duplication
 - Inversion

CHROMOSOME MUTATION



- Translocation
- Chromosomal mutations in germ-line cells in humans result in a substantial proportion of congenital abnormalities which can impact the offspring that are produced
- Chromosomal mutations in somatic cells may result in cell death, or loss/reduction of cell function which may contribute to cancer

3. SOMATIC MUTATIONS AND GERM-LINE MUTATIONS AND THEIR EFFECT

⇒ 3.1 Somatic mutations

- Mutations in the somatic or normal. Body cells that occur in individuals after conception
- They cannot be passed on to offspring
- The mutations are carried in the daughter cells of the original mutant cell and therefore, the initial impact is only on specific cells and tissues
- Somatic mutations can cause cancer and disease in organisms
- They do not directly change the allele frequency in the gene pool

⇒ 3.2 Germ-line mutations

- Germ line cells are the gametes that carry DNA and genes through fertilisation to the next generation zygote
- Mutations in germ-line cells result in them being carried to all cells in the offspring
- Inherited diseases and disorders are a result of mutations in germ-line cells
- They have the potential to change allele frequency

4. THE SIGNIFICANT OF 'CODING AND 'NON-CODING' DNA SEGMENTS IN THE PROCESS OF MUTATION

⇒ 4.1 Mutation in 'coding' DNA segments

- Coding sections of DNA segments are comprised of the genes that are templates for the formation of polypeptides
- If a mutation occurs in the coding, then the consequences can include
 - A different mRNA codon but the same amino acid (silent mutation)
 - A point mutation may result in a different amino acid (missense mutation)
 - Mutation may result in the substitution of a 'stop codon' (missense mutation) which terminates the synthesis of the rest of the polypeptide chain and hence the protein
 - Then becomes non-functional
- SILENT MUTATION
 - A base substitution mutation that results in the same amino acid in the resulting polypeptide and thus no observable impact
- MISSENSE MUTATION
 - A base substitution mutation that results in one changed amino acid in the polypeptide sequence and may or may not change the functioning of the resulting protein
- NONSENSE MUTATION
 - A mutation that results in a stop codon being prematurely inserted into a polypeptide, so its synthesis is terminated, and it becomes non-functional

⇒ 4.2 Mutation in 'non-coding' DNA segments

- 'Non-coding' DNA segments have a range of functions including gene expression
- If a 'non-coding DNA experiences a mutation in a promoter sequence, DNA polymerase may not be able to bind, thereby preventing the transcription of a polypeptide'

5. THE CAUSES OF GENETIC VARIATION RELATING TO THE PROCESSES OF FERTILISATION, MEIOSIS AND MUTATION

- Mutations are the ultimate source of new alleles and genetic change

	Fertilisation	Meiosis	Mutation
How this causes genetic variation	Allows new combinations of alleles	Random segregation causes new combinations of chromosomes and hence alleles Crossing over gives new combinations of alleles among chromosomes	Produces new alleles and changes in gene regulation and expression or chromosome number and arrangement
Impact on gene pool	Natural selection may play a role by increasing the chances of more 'fit' parents producing the gametes	The process in meiosis result in variation in combinations of alleles but not alleles frequency	Potential to change allele frequency in gene pool

- Fertilisation and meiosis are the key events that facilitate genetic variation in sexual reproduction
- In-vitro fertilisation is used, along with genetic screening to reduce the chance of genetic disease

6. THE EFFECT OF MUTATION, GENE FLOW AND GENETIC DRIFT ON THE GENE POOL OF POPULATIONS

- Diversity within a gene pool may result from population size, mutation, genetic drift, natural selection, diverse environments and population movements resulting in gene flow

- The effect of mutations, genetic drift and gene flow on the gene pool are better in small populations

Cause	Effect	Evaluation
MUTATION	Initially mutation increases the variety of alleles in the gene pool	Most significant cause of change to gene pool mutation impact may be positive, negative or neutral
GENETIC DRIFT	Random changes in allele frequency due to a range of factors in small populations random events might lead to increases or decreases of allele frequency	Least important and less likely to alter gene pool composition unless the population is very small
GENE FLOW	Migration into a population can increase the genetic diversity due to gene flow Conservation management practices include the maintenance of wildlife corridors to ensure gene flow	Generally positive impact by increasing the diversity of the gene pool

BIOTECHNOLOGY

INQUIRY QUESTION: How do genetic techniques affect Earth's biodiversity?

- INVESTIGATE THE USES AND APPLICATION OF BIOTECHNOLOGY (PAST, PRESENT AND FUTURE)

Applications	Past	Social implications	Ethical implications
agricultural uses and applications	<p>selective breeding, fermentation of food and beverages for food preservation and maintaining nutrition, simple aquaculture to improve availability of protein sources</p> <p>genetic modification of soybeans started in 1988 and is now the most widely used GMO</p> <p>biological controls were used to control some pests (e.g. in 1925 <i>Cactoblastis</i> moth larvae was used to control prickly pear in NSW and Queensland)</p>	<p>improvements in health and nutrition</p> <p>some social groups experienced improvements in productivity, wealth and increased leisure and autonomy</p> <p>countries responded differently to the acceptance of GM organisms as food or food ingredients and legislated and regulated accordingly</p>	<p>habitat destruction for many species, loss of biodiversity</p> <p>animal welfare (e.g. free-range versus penned animals, live animal transport and export)</p> <p>soil erosion, degradation and salinity</p> <p>irrigation impacts on river systems, increased salinity</p>
industrial and environmental uses and applications	<p>from 1920s bacterial sludge was used in sewage treatment</p> <p>anthrax was used in 2001 against 22 people in the USA in what is regarded as bioterrorism</p> <p>fermentation was used for production of useful substances such as citric and lactic acid</p> <p>in 1989 bacteria were used to help clean up the Exxon oil spill in Alaska</p>	<p>better waste disposal and reduced contamination of water and soil</p> <p>reduced security because of increased bioterrorism threat; the Biological Weapons Convention (1972) was co-signed by 103 nations but there has been substantial evidence that not all signatories have complied</p> <p>less dependence on imports, improved access to a range of products</p> <p>reduced damage from accidental environmental spills</p>	<p>international conventions require scrutiny and there is an increased need for nations to be defensively prepared for the use of biological weapons</p>
medical uses and applications	<p>production of vaccines, anti-venoms, antibiotics (e.g. penicillin), prebiotics and probiotics, dietary supplements and herbal remedies</p> <p>from 1920s insulin was extracted from pigs and cattle for the treatment of diabetes</p>	<p>need to ensure quality and efficacy of many herbal remedies and pharmaceuticals</p> <p>some supplements are not subject to regulation and labelling restrictions in some countries</p> <p>treatments are more safe and predictable</p>	<p>precedence set for the use of animal models for research and testing of products</p> <p>the discussion of benefits versus harm caused is raised in production of MAbs which are used to fight, diagnose and research diseases as well as to test new drugs</p>

⇒ 1.1 Past uses of biotechnology

⇒ 1.2 Present uses of biotechnology

- AGRICULTURAL USES AND APPLICATIONS
 - Transgenic crops counter abiotic and biotic factors
 - Crops with increased nutritional benefits increase vitamin content. This is achieved through selective breeding and genetic modification
 - RNAi has been used to reduce the function of three genes to retain higher levels of oleic acid
 - GM canola produces an oil resembling that from coconuts or palms
- INDUSTRIAL AND ENVIRONMENTAL USES AND APPLICATIONS

- Products and processes that degrade toxic or harmful chemicals and wastes by the use of bacteria or other microorganisms have been developed
- Diagnostic tests detect and eliminate the illegal use of endangered species
- Biofuels are made from plant materials and animal waste
- Biodegradable plastics are made from petrochemicals and can be broken down more quickly
- Bioplastics have been made from corn starch
- MEDICAL USES AND APPLICATIONS
 - Biopharming produces proteins, vaccines, hormones and blood clotting and blood thinning agents
 - RNAi gene silencing is used to improve the production of antibodies for treating human diseases

⇒ *1.3 Possible future uses of biotechnology*

- AGRICULTURAL USES AND APPLICATIONS
 - Isolation and insertion of genes has reduced the need for nitrogenous fertilisers
 - There has been an expansion in the ability to control the gender of animals
 - Marker-assisted breeding to breed hornless cattle in order to end the painful practice of dehorning
- INDUSTRIAL AND ENVIRONMENTAL USES
 - Climate change, food and energy security and waste disposal
 - Biomarkers act as environmental indicators assess the level of exposure and the impact of pollution or damage from pollutants
 - Possibility of eliminating some exotic pests
- MEDICAL APPLICATIONS
 - Cultures of human stem cells for drug trials
 - Development of virus free cells, tissues or organs
 - Elimination of certain infectious diseases
 - Recombinants antibodies

⇒ *1.4 The social implications of biotechnology*

- Possible impacts on developing countries through losing traditional markets and products
- Food safety and security
- Potential for environmental damage
- Loss of autonomy
- Potential for bioterrorism or the use of biological weapons in warfare
- Lab safety and security of genetically modified plants and animals
- Ethical concerns about future directions
- Less starvation and undernutrition
- Less environmental damage because of more efficient food production
- Increased life expectancy and quality of life

- Personalised medicine and management of health risks
- More resources and environmentally friendly and reliable energy supplies

⇒ 1.5 The ethical uses of biotechnology

- Use of animal parts
- Deliberately breeding animals for research of disease
- Change the genomes of other organisms and potentially changing ecosystems and biodiversity

⇒ 1.5 Researching future directions of the use of biotechnology

- Bio-computers which control gene expression being inserted into a living cell
- Gene therapy involves attempting to correct faulty genes in human cells

⇒ 1.6 Evaluating the changes to the Earth's biodiversity due to genetic techniques

- Tools of genetic research are included
- Covered in genetic technologies
- Initial increase in genetic diversity
- Uptake of the use of genetically identical or similar organisms
- Increase in rate of species extinction
- Increase in life expectancy and size of human population
- Elimination of species considered as pests to humans

⇒ 1.7 Evaluating the potential benefits for society of research using genetic technologies

- Less starvation and undernutrition
- Less environmental damage because of more efficient food production
- Increased life expectancy and quality of life, personalised medicine and management of health risks
- More resources and environmentally friendly and reliable energy supplies, including better waste management or elimination and less pollution

GENETIC TECHNOLOGIES

INQUIRY QUESTION: Does artificial manipulation of DNA have the potential to change the population forever?

1. INVESTIGATE THE USES AND ADVANTAGES OF CURRENT GENETIC TECHNOLOGIES THAT INCLUDE GENETIC CHANGE

Technology	Uses	Advantages
a knockout (KO) gene is an organism containing a gene that has been made inoperable	used to study gene function by comparing a knockout with normal organisms mice are frequently used as knockout animals because of similarities with humans	KO mice research has helped understand human diseases including cancer, obesity, heart disease, diabetes, arthritis, substance abuse, anxiety, aging and Parkinson's disease
RNA interference (RNAi) or gene silencing	RNAi technology can identify which genes are responsible for particular traits; plant breeders can then produce more productive but non-genetically modified plants researchers have used RNAi in chicken embryos, causing male to female sex reversal which assists understanding sex determination in animals in aquaculture the technology reduces stock losses due to diseases (e.g. developing an experimental anti-viral that prevents prawn mortality from Gill-associated virus)	a therapeutic agent used to control disease and prevent infection in plant and animal cells the CSIRO claims the process is fast, reliable, flexible, stable and controllable; the gene editing is temporary ability to produce plants with desirable properties without using GM so RNAi is more acceptable to many people in aquaculture, more sustainable feed formulations, enhanced growth, better survival and feed conversion rates and increased tolerance to viral diseases
marker-assisted breeding	use of genetic markers helps select organisms for selective breeding its use, combined with viral screening technology, has helped the tiger prawn aquaculture industry to develop breeds highly tolerant to endemic diseases	speeds up conventional selective breeding has increased pond yields of black tiger prawns by over 50%, easing pressure on wild stocks and making prawn aquaculture more sustainable
chromosome engineering, including manipulating chromosomes, results in rearrangements such as insertions, deletions and translocations also includes manipulation of meiosis, resulting in alterations in numbers of whole chromosomes or homologous chromosome sets	used to improve crop species such as corn, soybeans, rice, barley and potatoes	enables research into some diseases (e.g. myeloid tumours associated with chromosomal abnormalities)
microbial vectors involve microinjection using Ti plasmids and microprojectile bombardment	responsible for the majority of GE plants in commercial production although successful in fruits and vegetables, the Ti plasmid has generated limited success in grain crops though modification of the process has resulted in recent success in rice and sugar cane microprojectile bombardment involves delivering naked DNA to plant cells by 'shooting' them with microscopic pellets to which DNA has been adhered; this is effective in corn, rice and other cereal grains	overcame the difficulty of introducing cloned genes through cell walls and membranes into plant cells
genetic screening combined with in-vitro fertilisation (IVF)	embryos may be screened and selected to avoid inherited disease there has recently been the case of a human produced from three genetic parents where the maternal DNA (nucleus) was inserted into an enucleated egg from another female to avoid an inherited disease carried on the mother's mitochondria	harmful germ-line mutations can be avoided in offspring

2. COMPARE THE PROCESSES AND OUTCOMES OF REPRODUCTIVE TECHNOLOGIES

⇒ 2.1 Artificial insemination

- Collection, storage and transport of semen from male animals with desirable qualities used to fertilise a large number of females
- Large numbers of more desirable animals
- Less genetic diversity
- Global benefits from storage and transport of semen

⇒ 2.2 Artificial pollination

- Involves the transfer of pollen to stigmas to enable pollination and fertilisation
- Can be used to deliberately produce hybrids or for selective breeding
- Natural pollinations need to be prevented by physically removing stamens or using other techniques to induce male sterility
- Improved plant cultivars such as rice and many types of plant hybrids with hybrid vigour

3. INVESTIGATE AND ASSESS THE EFFECTIVENESS OF CLONING

⇒ 3.1 Whole organism cloning

- Produces multiple identical copies of organisms with selected characteristics
- Plant cloning is used to produce large numbers of the desired plant
 - Tissue culture is the predominant method used here
- Animal cloning occurs in two procedures
 - Embryo splitting results in identical cells that develop into genetically identical individuals. The qualities of the embryo are often unknown
 - Somatic cell nuclear transfer (SCNT) uses genetic material from mature animals to result in animals with identical genomes
- Effectiveness is:
 - High pregnancy losses and high morbidity and mortality during neonatal period
 - May preserve endangered species
 - Cloning is expensive and prone to failure
 - Surrogate mothers suffer increased health risks
 - Used cloned animals for food remains

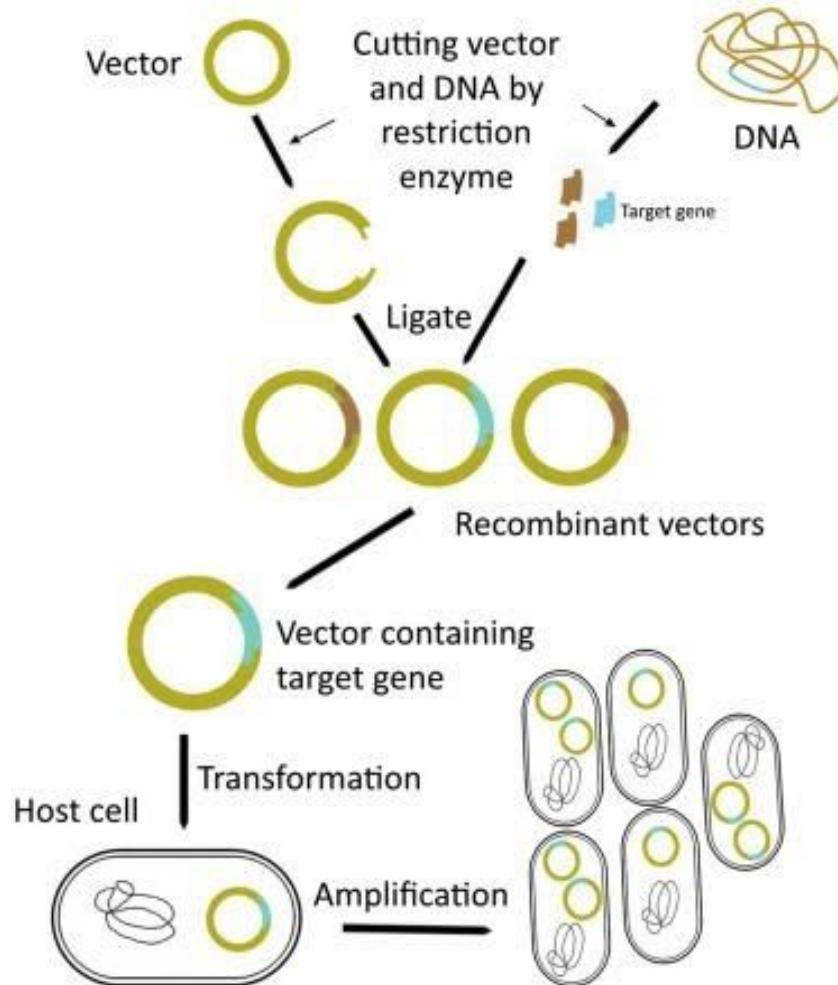
⇒ 3.2 Gene cloning

- Uses vector plasmid results in multiple copied of genes usually in bacterium such as E. coli
- Genes that have been expressed can be cloned
- The polymerase chain reaction PCR is one method of copying DNA or genes
- It is fast and efficient and allows high throughput
- It is useful when there are only small amounts of the required gene/DNA

4. DESCRIBE TECHNOLOGIES AND APPLICATIONS USED IN RECOMBINANT DNA TECHNOLOGY

⇒ *4.1 Recombinant DNA technology*

- Involved gene-shuffling
- A specific segment of DNA is isolated and removed from one organism and attached to another piece of DNA, often from a different organism
- Produces new characteristics in the host organism or enables the host organism to produce specific substances



- Essential steps
 - Gene of interest is identified and the DNA containing the gene is extracted and purified
 - Cutting and joining the DNA is done by using a restriction enzyme which results in a single piece of DNA containing DNA from two different species joined together
 - Monitoring the cutting and joining – electrophoresis can be used to separate DNA fragments
 - The recombinant DNA is amplified or modified

- Transforming hosts, such as bacteria with the recombinant DNA allows the genes to be carried into nuclear DNA in multicellular organisms

5. EVALUATE THE BENEFITS OF USING GENETIC TECHNOLOGIES IN AGRICULTURAL, MEDICAL AND INDUSTRIAL APPLICATIONS

⇒ **5.1 Agriculture**

- The ability to more rapidly, precisely and cheaply breed organisms that are more suitable for growth in changing environments; increased salinity, increased drought frequency or exposure to new diseases
- The ability to produce more nutritious foods
- Need for less or more effective use of pesticides
- Less use of labour because of a reduced need for cultivation
- Ability to raise livestock more humanely
- Ability to control the gender of animals in order to increase productivity and eliminate exotic pests

⇒ **5.2 Medicine**

- GM vaccines are safer and no longer made from denatured pathogens which occasionally cause disease
- Genetic engineering has been used to mass-produce insulin, human growth hormones, follistim, human albumin, monoclonal antibodies, blood clotting and thinning factors, vaccines and many other drugs
- Better analytical techniques can be used to detect substances such as specific proteins in cell and tissues samples
- Discovering the functions of certain genes has led to a better understanding of some diseases
- Cancer-treatment peptides can stimulate the ability of the immune system to recognise the difference between self and non-self-resulting in more effective treatments

⇒ **5.3 Industry**

- Mass quantities of proteins
- Gene technologies allow the development of highly sensitive analytical procedures
- Processes and products that would be difficult to develop using traditional biotechnological techniques
- Ability to produce renewable and better fuels and products to clean up existing pollution
- Biodegradable plastics

6. EVALUATE THE EFFECT ON BIODIVERSITY OF USING BIOTECHNOLOGY IN AGRICULTURE

7. INTERPRET A RANGE OF SECONDARY SOURCES TO ASSES THE INFLUENCE OF SOCIAL, ECONOMIC AND CULTURAL CONTEXTS ON A RANGE OF BIOTECHNOLOGIES

INFECTIOUS DISEASES

CAUSES OF INFECTIOUS DISEASES

INQUIRY QUESTION: How are diseases transmitted?

- Disease
 - Any condition that impairs the normal living processes and is recognised by the presence of specific symptoms.
 - Has adverse effects on the normal functioning of a living thing or parts of a living thing
 - Can vary significantly in cause, severity and symptoms they cause
- Infectious diseases
 - Have a causative agent or infective agent

- Known as **A PATHOGEN**
- They are usually contagious
- Caused by the invasion of the body by a disease-causing agent
- Non-infectious diseases
 - Caused by genetics, environment influences or cellular malfunctions
 - Multiple factors contribute to the development of non-infectious diseases
 - They do not involve a pathogen and are not contagious
- Communicable disease
 - Word used to describe a disease than can be transmitted from plant to plant or from animal to animal.

1. INFECTIOUS DISEASES CAUSED BY PATHOGENS

⇒ *1.1 Pathogens: microorganisms, macro-organisms and noncellular*

- Infection is the presence of a disease-causing organism in or on the body of a host
- Infectious diseases are caused by invasion by a pathogen and can be transmitted from one host to another

⇒ *1.2 Classifying different pathogens that cause disease in plants and animals*

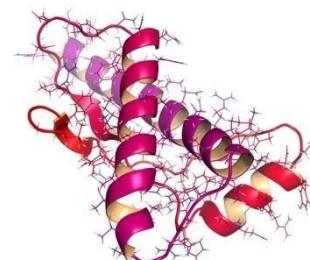
- Pathogens can be prions, bacteria, viruses, protozoa or protists, fungi or macroscopic parasitic animals
- MICROSCOPIC: bacteria, viruses, fungi and protozoans
- MACROSCOPIC: parasites like arthropods, flatworms and roundworms

Type of Pathogen	Some Distinguishing Features
Prion	Defective form of a protein molecule; does not contain DNA or RNA; mostly attacks brain or nerve cells
Virus	Non-cellular; contains DNA, RNA and protein coat; requires a living host cell to replicate
Bacteria	Prokaryotic cell; divides quickly and/or produces toxins
Protozoan	Eukaryotic cell (single-celled organism); may have a complex life cycle
Fungi	Eukaryotic cell with cell wall; spreads via spores or rapid division; some infect

	external skin and nails while others enter the host's body
Macroparasites	Eukaryotic cell with cell wall; spreads via spores or rapid division; some infect external skin and nails, while others enter the body

PRIONS

- Infectious agents that consist only of protein; no nucleic acid therefore non-cellular
- Able to induce abnormal folding of specific normal cellular proteins known as prion proteins
- Prion diseases are also called transmissible spongiform
- encephalopathies
- Creutzfeldt-Jakob disease (CJD)



VIRUSES

- Microscopic pathogens from 20-40 nanometres
- Is small piece of genetic material encased in a shell called a capsid
- Referred to as non-cellular pathogens
- Can infect all types of living things
- Zika Virus



BACTERIA

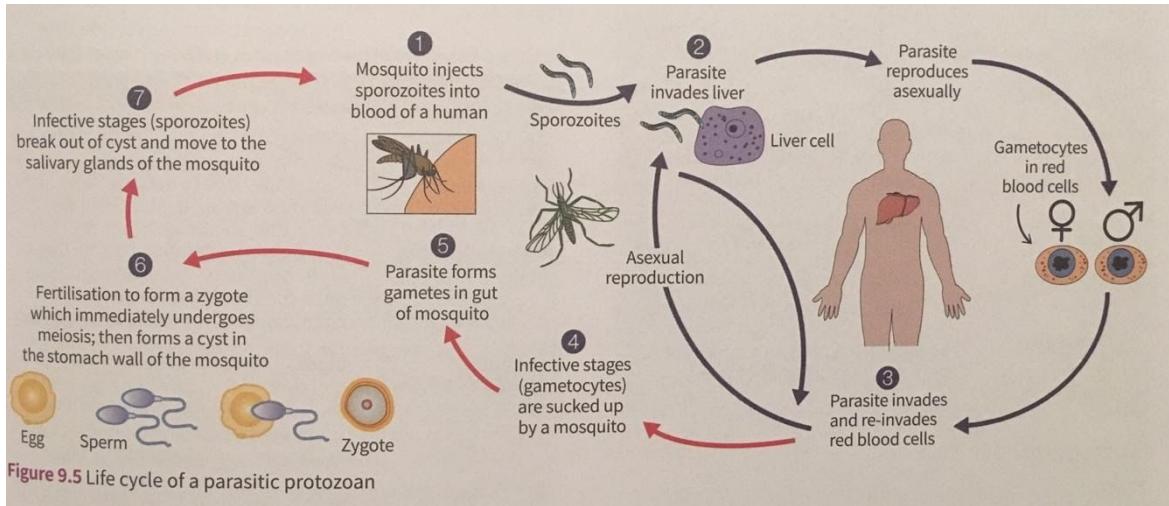
- Prokaryotic, unicellular microorganisms, DNA in a single loop
- 1% are disease causing
- 1000 nanometres in size
- Bordatella pertussis is a bacterium that causes whooping cough



PROTOZOANS

- Eukaryotic microorganisms, larger than bacteria, DNA in nucleus
- Most are less than 50 micrometres

- Parasitic protozoans have complex life cycles involving several hosts
- Malaria from mosquito *plasmodium falciparum*



FUNGI

- Fungi are eukaryotic organisms
- Yeasts, moulds and crop-destroying rusts and smuts
- Many fungi are beneficial and play a key role as decomposers in ecosystems and in the fermentation process however some cause disease

MACROPARASITES

- Microorganisms that can cause diseases
- Ticks, leeches, flees, tapeworms
- Have complex lifecycles



⇒ 1.3 Modes of transmission of infectious diseases

- Spread involves a wide range of interrelated factors
- Transmission must occur for an infection to spread

DIRECT CONTACT

- Direct contact between people (touch, saliva, mucus, contaminated blood or other bodily fluids)
- Individuals physically transferring the pathogens, e.g. through touching, kissing or having sexual intercourse
- Direct droplet transmission occurs when someone sneezes or coughs and small droplets of mucus which may contain pathogens are ejected as fine spray



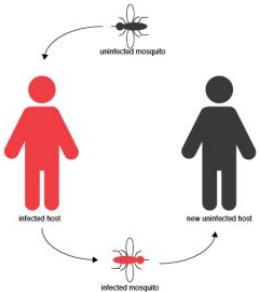
INDIRECT CONTACT

- Airborne transmission of droplets; respiratory pathogens are usually transmitted through the air
- Transmission from fomites (inanimate objects that become contaminated)
- Transmission from contaminated food and drinking water
- Transmission from an infected animal
- Involves objects being contaminated with pathogens, e.g. a used tissue, a fork with saliva traces, used bedsheets, or contaminated medical equipment



VECTOR TRANSMISSION

- Can be mechanical or biological
- Mechanical involves an animal carrying a pathogen from one host to another without being infected itself
- Biological transmission includes insects and other arthropods
- Spread of pathogens by contaminated air, food and water
- Involves animals assisting in the transfer of pathogens between individuals
- *Biological vectors*, mosquitos, transfer the pathogen from one individual to another
- *Mechanical vectors* such as flies, physically transfer the pathogen from one person to another without being infected themselves



Sarah gets the cold sore virus from kissing her aunt	Direct contact
Dylan goes bush walking and gets a tick bite. This tick transmits bacteria into him that cause Lyme disease	Vector transmission
Judy catches influenza from a man sitting beside her on the train	Direct contact
A body piercing salon accidentally reuses a needle, spreading hepatitis C virus from one client to the next	Indirect contact

Jodi eats chicken that is slightly raw and gets salmonella poisoning	Vehicle transmission
Alyssa gets bacterial conjunctivitis (eye infection) after playing with a contaminated doll at preschool	Indirect contact
Nate has unprotected oral sex and as a result gets syphilis, a bacterial sexually transmitted infection	Direct contact
Pip and John share a drink bottle, which also results in them sharing the glandular fever virus	Direct contact
James drinks tap water in South America and gets giardia	Vehicle transmission
Ivan gets bitten by a mosquito in the tropics and develops malaria	Vector transmission
Jan eats contaminated beef and gets Creutzfeldt-Jacob disease	Vehicle transmission
Andrew had diarrhoea after touching a contaminated door knob	Indirect contact

⇒ *1.4 Investigating the transmission of a disease during an epidemic*

- An epidemic is an outbreak or unusually high occurrence of a disease in a population or region
 - It is the rapid spread of a infectious disease to a large number of people in a particular population in a relatively short period of time
 - Affect those in the population who do not have an acquired or inherent immunity
 - When it spreads to the entire nation or other countries it is called a pandemic
 - A pandemic is an outbreak of a disease over a whole country or on a global scale
 - Epidemics and pandemics are becoming more prevalent
- TRANSMISSION

- It is usually rapid
- There are many factors that increase exposure to pathogens and cause the spread of disease
- Continual source of a disease is known as a reservoir (infected person during AIDS epidemic, rats and fleas during the Black Death, contaminated water in cholera and typhus epidemics)
- In dense areas (cities) pathogens can spread more easily and rapidly, humans are in close proximity with each other and livestock
- Social and economic factors like contaminated water supply and poor sanitation facilities
- Environmental factors (change in climate), e.g. ticks in Africa, Asia and southwest Europe spread due to global warming
- Loss of healthcare systems and shortages of medical supplies (civil war)
- Natural disasters (heavy rain, mudslides, cyclones, earthquakes and flooding) damage sewerage infrastructure, causes contamination or water
- Viruses can evolve and spread faster
- Globalisation (global trade, migration and global travel) help spread disease
- Change in susceptibility

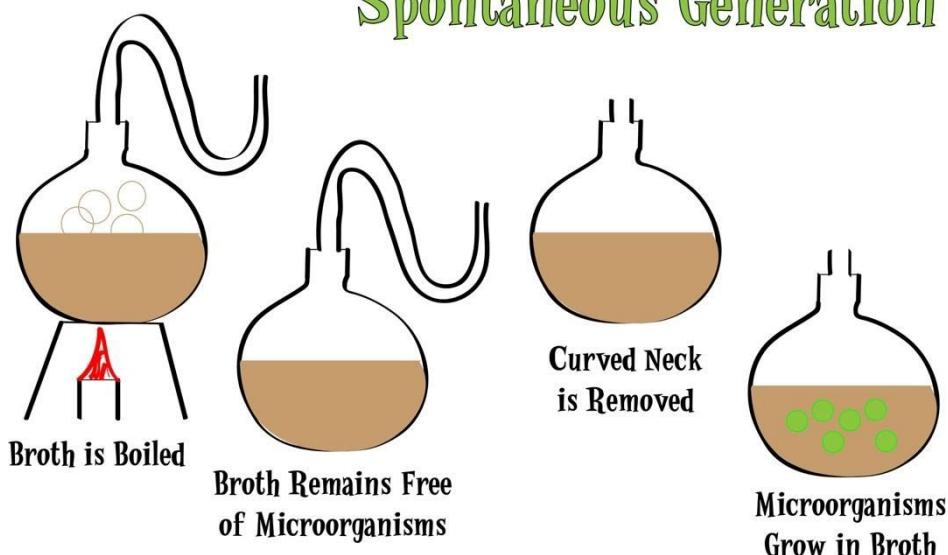
YEAR	DISEASE	REGION	NUMBER OF DEATHS
541-2	Bubonic plague	Europe and Asia	25-50 million
1346-53	Black death	Europe, Asia, Africa	72-200 million
1545 and 1576	Smallpox	Mexico	17 million
1665-66	Bubonic plague	Europe	100 000
1817-24	Cholera pandemic	Asia, Europe	>100 000
1918-20	Spanish flu	Worldwide	20-50 million
1980s	HIV	Worldwide	35 million
2003	SARS	Asia and Canada	800 deaths
2013-16	Ebola	West Africa	>11 300 deaths
2017	Plague	Madagascar	207

2. INVESTIGATING THE WORK OF ROBERT KOCH AND LOUIS PASTEUR

⇒ 2.1 Pasteur's experiment on microbial contamination

- It used to be believed that living matter (moulds, maggots) could generate spontaneously also known as spontaneous generation from non-living matter
- Louis Pasteur discovered the microbes (bacteria and moulds) can cause contamination and disease
- He demonstrated this with his swan-neck flask experiment disproving spontaneous generation

Pasteur's Test of Spontaneous Generation



- Pasteur discovered that microbes were responsible for the souring of alcohol and developed the process called pasteurisation
- It led to the development of vaccinations

FIRSTHAND INVESTIGATION

Modelling Pasteur's experiment

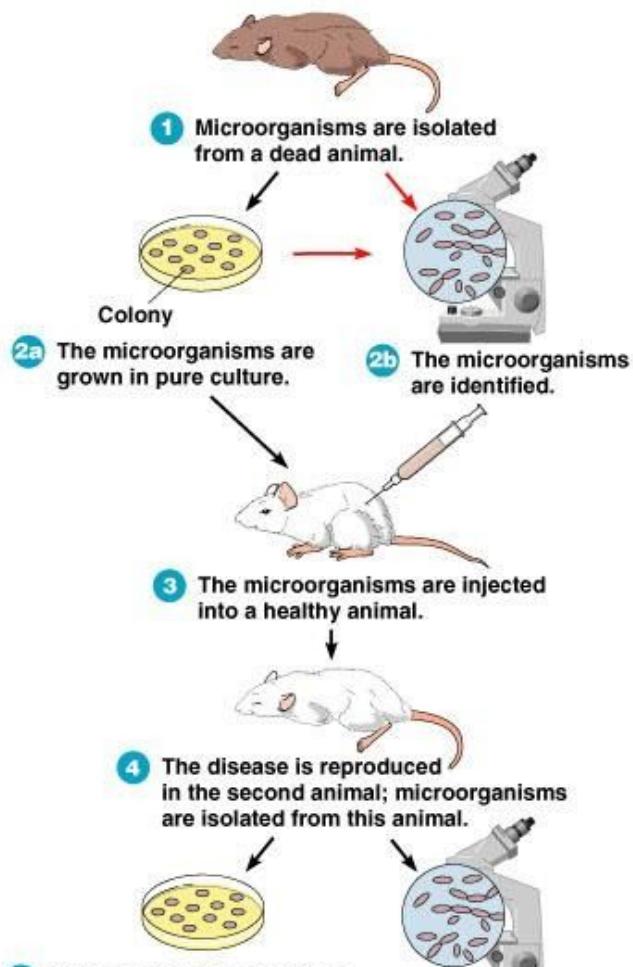
Aim: to model Pasteur's experiment to identify the role of microbes in contamination.

Method:

- Dissolve a meat extract cube in hot water.
- Set up the equipment with a conical flask fitted with a one-holed stopper through which a glass tube. Bent into an S-shape is passed to prevent entry of air.
- Set up an identical flask and stopper but containing a straight piece of glass tube.
- Place equal amounts of broth in both flasks and gently boil for 15 minutes.
- Observe for several weeks, looking for changes in broth colour and, if safe to do so, odour. Follow safe procedures when investigating the contaminated liquid.
- Safely dispose of contaminated liquid by autoclaving.

⇒ 2.2 Koch's postulates

- Koch showed that bacteria were the cause of a disease called anthrax
- He designed four rules of procedure for showing that a particular microorganism is the cause of a particular disease
 - 1. It must be shown that the microorganism believed to be the cause of the disease is always present in the diseased organisms
 - 2. Microorganism must be isolated and grown in a pure culture; that is, a. culture only containing that microorganism
 - 3. Microorganisms from the pure culture, when injected into a healthy organism without the disease, must produce the disease
 - 4. Microorganisms isolated from the experimental organisms, grown in pure culture, and compared with the microorganisms in the original culture, are shown to be identical



3. THE CAUSES AND EFFECTS OF DISEASES ON AGRICULTURAL PRODUCTION

⇒ 3.1 Animal diseases

DISEASE	CAUSE	EFFECT
Sheep lice	Insect: <i>Bovicola ovis</i>	Sheep lice costs producer's inn NSW >\$100 million per year in lost production and treatment costs
Anthrax I humans, cattle, sheep, horses, goats and deer	Bacterium: <i>Bacillus anthracis</i>	Passed on through the inhalation of spores which have been known to survive for 70 years. Has a mortality rate of 50% to 80%
Newcastle disease. In fowls, turkeys, pigeons, parrots and other domestic and wild birds	Virus: <i>Avian. Pneumoencephalitis</i>	The disease is highly contagious
Foot-and-mouth disease in cattle, water buffalo, sheep, goats, pigs, antelope, bison and deer	Virus with seven serotypes: A, O, C SAT1, SAT2, SAT3 and Asia 1, and over 60 strains	Caused losses of \$19 billion. It is highly contagious

⇒ 3.2 Plant diseases

- Plant crops are often staple foods
- Losses from diseases are particularly damaging

DISEASE	CAUSE	EFFECT
Potato blight	Fungi	Caused 1 million deaths from starvation
Golden potato cyst nematode	<i>Globodera rostochiensis</i>	Crop damage was a serious threat to Australia's potato industry
Blast and bacterial blight in rice	<i>Blast fungus</i>	

Stem rust and leaf blotch in wheat stem rust in barley	Stem rust in wheat and barley	Caused losses of \$19 billion. It is highly contagious
Leaf blight and bacterial. Stalk rot. In maize	Leaf blotch	Altogether these 3 diseases caused half of the population cereal food grain loss *below*
Grain smut in sorghum other domestic and wild birds		
Foot-and-mouth disease in cattle, water buffalo, sheep, goats, pigs, antelope, bison and deer	Virus with seven serotypes: A, O, C SAT1, SAT2, SAT3 and Asia 1, and over 60 strains	

4. THE ADAPTATIONS OF DIFFERENT PATHOGENS THAT FACILITATE THEIR ENTRY INTO AND TRANSMISSION BETWEEN HOSTS

⇒ *4.1 Adaptations of different pathogens that facilitate their entry into hosts*

- Pathogens have adaptations that facilitate their entry into cells and tissues and their transmission between hosts
- Pathogens have elaborate ways of adaptation
- In plants, protective barriers include physical barriers
 - Thick cell walls
 - Thick cuticles
 - Presence of a secondary cell wall or thick bark
- Plants also have chemical barriers
 - Antimicrobial compounds
- In animals, protective barriers include physical barriers
 - Thick, fairly tough covering of skin
 - Epithelial surfaces
- Animals also have chemical barriers
 - Include PH of skin and other body fluids

- Many pathogens have adaptations for overcoming the protective barriers and the host's cleaning mechanisms
 - Some bacteria produce chemicals that bind them to the host tissues
 - Once they have gained access, many pathogens infect hosts without entering the host's cells. These are called extracellular pathogens.
 - Intracellular pathogens have adaptations to facilitate their entry into the host cell, survival and replication within the cell, and exit from the cell
 - Intracellular pathogens have acquired genes that encode proteins which interact with particular molecules of the host cell

⇒ *4.2 Adaptations of different pathogens that facilitate their entry between hosts*

- All pathogens have a mechanism of transfer from one host to another or they will die if the host dies
- Diseases that cause diarrhoea provide an example of adaptations that facilitate transmission
- Some pathogens alter the behaviour of the host to facilitate their transmission

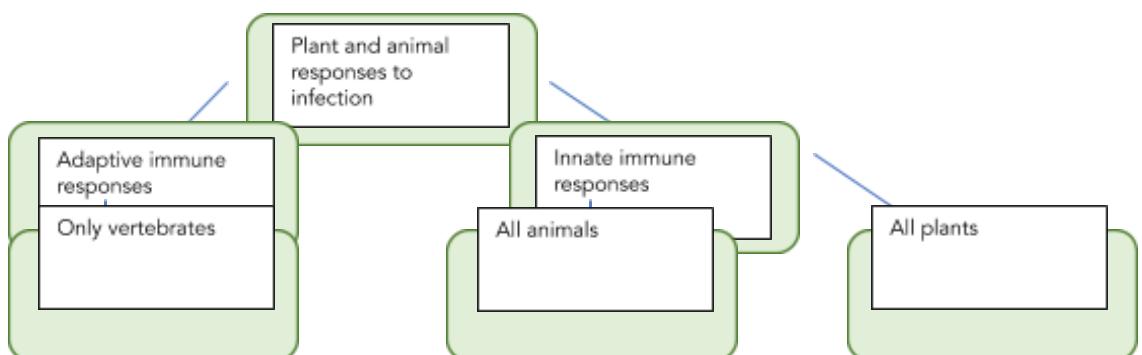
RESPONSES TO PATHOGENS

INQUIRY QUESTION: How does a plant or animal respond to infection?

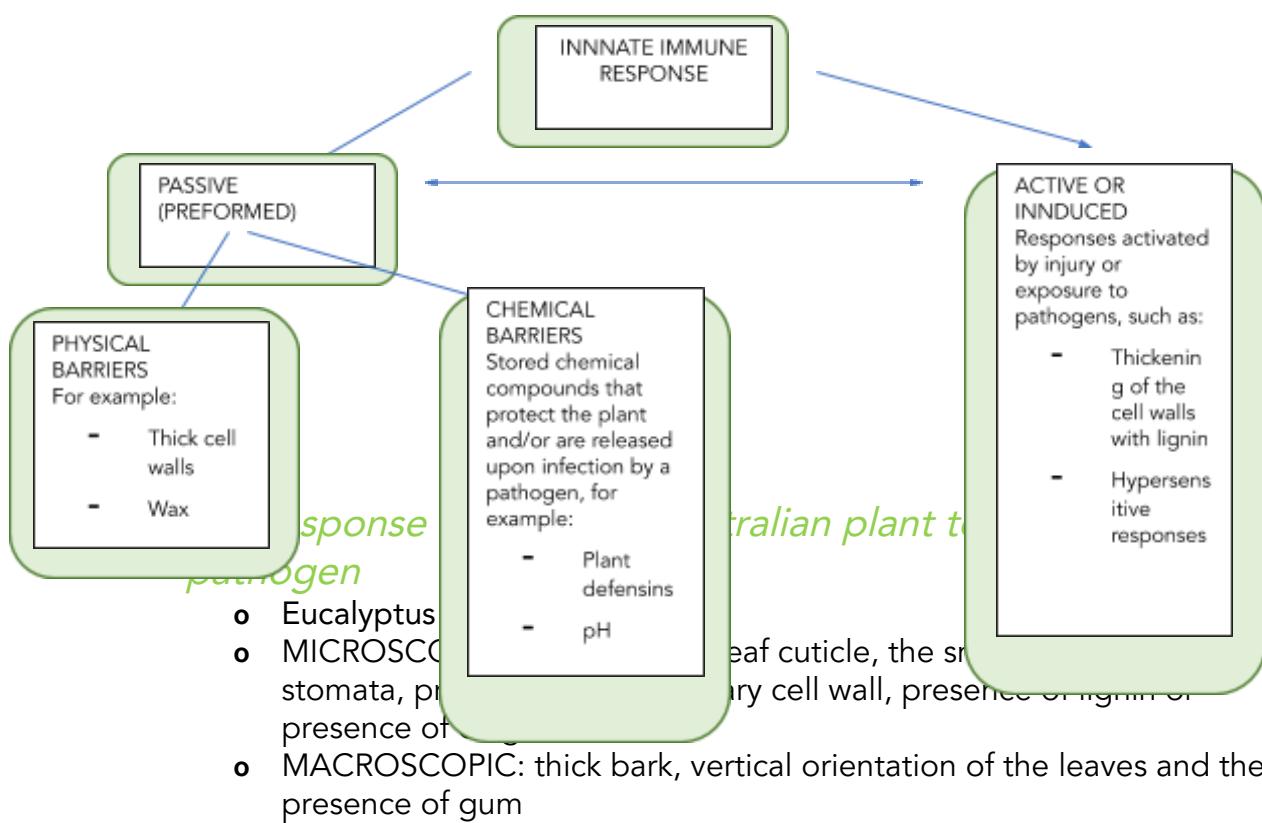
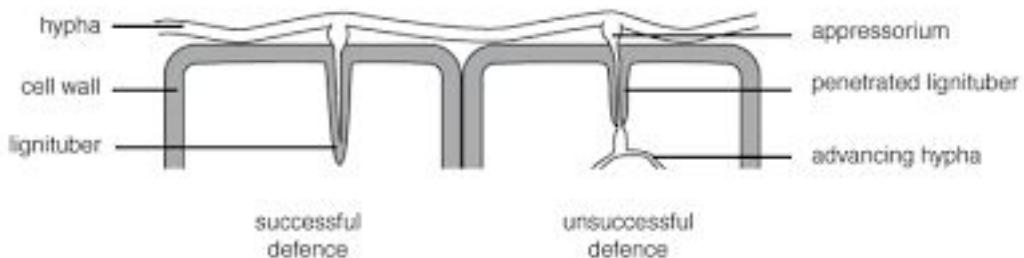
1. RESPONSES OF PLANTS TO PATHOGENS

⇒ *1.1 Fungal and viral pathogens*

- Viral diseases are common amongst agricultural crops
- Fungal diseases are common in Australian plants
- Can include mould, rust, ink spots and powdery mildew
- Plants are animal responses are summarised below



- Infection is a battle between the invading pathogen and the host
- Plants must be able to protect themselves against a wide range of pathogens
- Plants can only rely on preformed defences and innate immune responses
- PLANTS DO NOT HAVE AN ADAPTIVE IMMUNE SYSTEM



PASSIVE DEFENCES (PREFORMED)	ACTIVE DEFENCES OR INDUCED RESPONSES
PHYSICAL BARRIERS / CHEMICAL BARRIERS Thick cuticle Stored oils function as preformed chemical defences, not only	The formation of barrier zones in the new tissue produced by the vascular cambium; these zones protect the healthy sapwood from damage by separating it from the adjacent infected or damaged tissues; in some

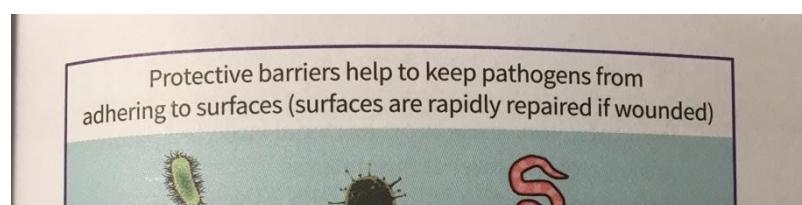
	against defoliating animals but as antifungal and antibacterial agents or for priming of defences in both the host and neighbouring plants	Eucalyptus these barrier zones contain gum The development of periderm , which separates damaged tissue from the healthy tissue and prevents the spread of the disease(s)
Thick bark		Secretion of protective gums that seal the wound from further infection
Waxy leaves		
Dry leaf structures Vertically hanging leaves prevent the formation of moisture on the leaf which provides some protection from water moulds		

- Range of pathogens pose a threat to a range of Eucalyptus in Australia
- Myrtle rust pathogen and root rot are common
- Root rot pathogen: *Phytophthora cinnamomi* is the named fungal infection
 - It is a soil borne water mould that produces a disease called root rot
 - Causes severe damage to native ecosystems including *Eucalyptus marginata*
 - The tree's responses include
 - Mass production of defence proteins
 - Activation of defence pathways that involve the production of a number of defence hormones
 - Hypersensitive response or rapid cell death near or at the invasion site
 - Lignin synthesis to reinforce physical barriers such as cell walls

2. RESPONSES OF ANIMALS TO PATHOGENS

⇒ *2.1 Innate immune defences and responses of animals to pathogens*

- Include:
 - Preformed defences to help prevent pathogens from entering
 - Recognition of the presence of a pathogen
 - Immune responses that are rapidly activated when the pathogen crosses protective barriers and begins to replicate



- At the tissue level
 - PHYSICAL barriers external and internal epithelial (the skin)
 - Thick, dry and scaly skin, the closed surface of unbroken skin and mucous membranes
 - CHEMICAL barriers at the tissue level include antimicrobial substances
 - Antibacterial enzyme secreted in saliva and tears, acid pH of the stomach and digestive enzymes, antibacterial and antifungal peptides produced in the lower intestinal tract
- Most epithelial surfaces also contain normal body flora that compete with pathogens for nutrients and attachment sites on cells
- At the cellular level:
 - PHYSICAL barriers include cell membrane, nuclear envelope and compartmental borders
 - CHEMICAL barriers include antimicrobial peptides in the plasma membrane cells, and lysosomes contain enzymes that have a number of functions
- If a pathogen crosses the barriers and begins to replicate, it is recognised as non-self
- In animals, pathogens activate the inflammatory response and phagocytosis
- INFLAMMATORY is the inflammation of the tissues in response to an infection; designed to isolate and destroy the foreign particles, prepares the tissue for healing
- PHAGOCYTOSIS the active process in which phagocytic cells enclose a pathogen and digest it in their interior
- Both responses bring about a physical and chemical change in cells and tissues

⇒ *2.2 Physical and chemical changes in response to pathogens*

- When a pathogen replicates it is recognised by phagocytes called macrophages
- MACROPHAGES are white blood cells found in tissues and in especially large numbers in the gastrointestinal tract, lungs, liver and spleen
 - They recognise and ingest and destroy the pathogens
- Macrophages and neutrophils produce toxic chemicals that help kill the engulfed microorganism
- Cytokines and histamines set up the inflammatory response
- Inflammatory response has three key roles in fighting infection

- Bring plasma proteins and phagocytes to the site of infection to help kill pathogen
- Provide a physical barrier to prevent further spread of infection and make the host aware of infection
- Promote the repair of damaged tissue
- o Physical and chemical changes in the cells and tissues as a result of inflammatory response include
 - Increased in blood vessel diameter, reduction in velocity of blood flow
 - Activation of cells lining the blood vessels to secrete adhesion chemicals which help the circulating white blood cells bind to infection site
 - Increased permeability of blood vessels which allows defence proteins to move out of the bloodstream
 - Fever and increase of temperature which kills bacterial and viral pathogens which thrive in low temperatures
- o Activation of endothelial cells to secrete proteins that bring about blood clotting

IMMUNITY

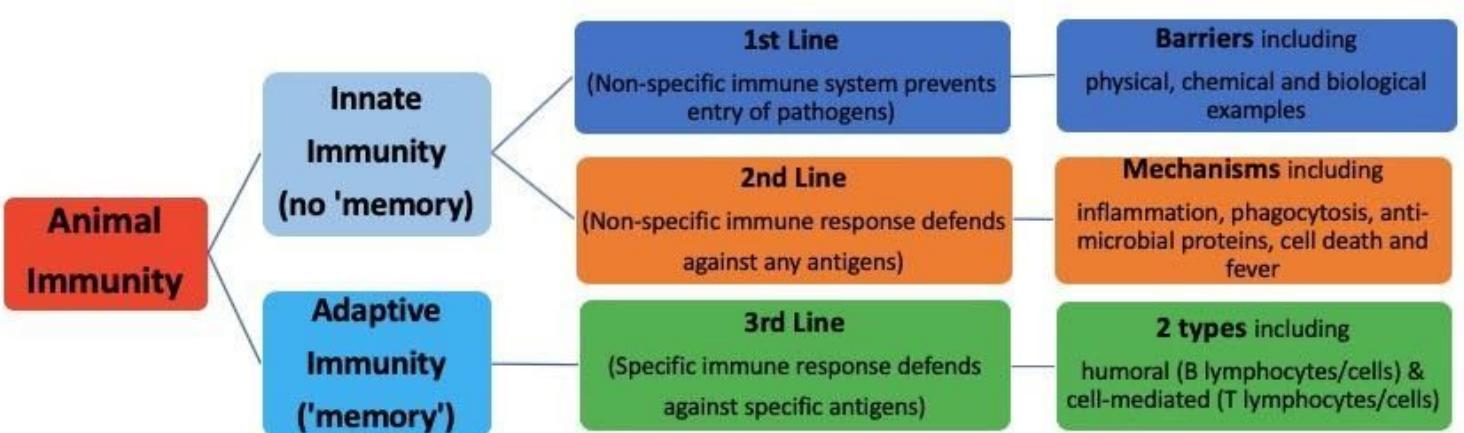
INQUIRY QUESTION: How does the human immune system respond to exposure to a pathogen

1. INNATE AND ADAPTIVE IMMUNE SYSTEMS IN THE HUMAN BODY

⇒ *1.1 Human immune systems*

- o When a non-self is in the body, the body responds with either. The innate immune system or the adaptive immune system

- o Both have special defence cells that are involved in eliminating pathogens
- o Both systems work together to create a complex response



⇒ **1.2 The innate immune system in the human body**

- o Main role is to prevent pathogens from entering and spreading in the body
- o Provides a front line of defence
- o Can fight different pathogens time and time again
- o Innate immune system is made up of;
 - The skin
 - Mucous membranes
 - Defence cells such as phagocytes
 - Various substances (cytokines and antimicrobial)
 - Complement system (system of soluble proteins that activate one and another in a chain reaction to support defence mechanisms)
 - Natural killer (NK) cells (cells that specialise in identifying cells that are infected)
- o Has no memory of the pathogen
- o Involved in the first line and the second line of defence
- o 1ST LINE
 - Non-specific immune system prevents entry of pathogens
 - Physical, chemical and biological barriers
- o 2ND LINE
 - Non-specific immune response defends against any antigens
 - Inflammation, phagocytosis, anti-microbial proteins, cell death and fever
- o PHAGOCYTOSIS
 - Two main types: neutrophils and macrophages
 - Phagocyte is able to engulf microbes via endocytosis
 - Chemicals and enzymes from a lysosome
- o INFAMMATORY RESPONSE

- Injuries to body tissue release histamines and prostaglandins that trigger an inflammatory response
- Chemicals cause increased blood flow to the site
- Side effects include localised redness, heat and swelling
- o SECRETION OF ANTIMICROBIAL PROTEINS
 - Lysosomes directly attack bacterial antigens by breaking down their cells walls
 - Proteins inn complement system cause the cells of invading microbes to burst
 - Interferon is involved in the immune response to viruses

⇒ ***1.3 Adaptive immune system in the human body***

- o Comes in if the innate immune system fails
- o It targets the pathogen more accurately
- o Can remember the pathogen and acts specifically on certain antigens
- o Adaptive immune system is made up of antibodies, B cells and T cells
- o ANTIBODIES
 - Each antigen has a specific shape which locks onto the specific shape of the antigen
 - Antibodies destroy the antigen which is then engulfed and digested by macrophages
 - Antibodies neutralise the pathogen by attaching onto the surface, activate the complement system or active other defence cells
 - They also activate the complement system
 - Some are formed through IgG, IgM, IgA or IgE
- o B CELLS - B LYMPHOCYTES
 - Develop in the bone marrow
 - Accumulate in the spleen and lymph nodes and circulate in the blood and lymphatic systems
 - Presence of an antigen stimulates the B cells to begin to divide to produce more B cells and specialist cells, called plasma cells which make large amounts of the specialised antibody and release them into the blood
- o T CELLS – T LYMPHOCYTES
 - They destroy the antigens directly
 - They do not generate antibody-producing plasma cells
 - Migrate from the bone marrow and develop in the thymus
 - They enter the bloodstream and are carried to lymph nodes and the spleen
- o HUMORAL AND CELL-MEDIATED IMMUNE RESPONSES
 - Antibodies are made available outside the cells in the blood and body fluids
 - This is the humoral response
 - The humoral response involves B cells that recognise antigens and pathogens
 - Antigen binds to B cell
 - Interleukins or helper T cells activate B cells

- B cells proliferate and produce plasma cells which make antibodies
 - B cells produce memory cells
- Cell mediated responses do not involve antibodies
- It involves the activation of T lymphocytes and the release of various cytokines in response to an antigen
 - Antigens bind to T cells
 - Interleukins activate T cell function
 - T cells proliferate, producing cytotoxic T cells which destroy the antigens

2. HOW THE IMMUNE SYSTEM RESPONDS AFTER PRIMARY EXPOSURE TO A PATHOGEN, INNATE AND ACQUIRED IMMUNITY

- Innate immunity is naturally present, and some people are naturally more resistant to specific diseases. It does not involve a response to an antigen and does not confer long-term immunity
- Acquired immunity requires exposure to a pathogen
 - It remembers the antigen because it produces memory cells
 - After first contact with the pathogens (antigen) it takes several days for the adaptive immune system to respond
 - After second infection with the same pathogen the adaptive immune system will recognise it and immediately release the antibodies needed to fight it

PREVENTION, TREATMENT AND CONTROL

INQUIRY QUESTION: How can the spread of infectious disease be controlled?

1. RANGE OF INTERALLATED FACTORS INVOLVED IN LIMITING LOCAL, REGIONAL AND GLOBAL SPREAD OF AN INFECTIOUS DISEASE

- How is the disease being transmitted
 - Direct

- Indirect
- Vector
- Etc
- o Where does the virus replicate
 - Location in the body e.g. upper respiratory tract
- o Type of setting where spread is most likely to occur
 - Poor hygiene practices or closed in environments
- o Individual susceptibility to the disease
 - Children and the elderly
 - Already have conditions
- o Transmission at different stages of infection
 - Most infectious period e.g. 3 days
- o Seasonality of infection
 - Seasonal patterns e.g. winter
 - Spreads in certain seasonal conditions

LOCAL	REGIONAL	GLOBAL
Limiting person-to-person transmission through personal hygiene	Limiting person-to-person transmission through border protection and entry screening	Limiting person-to-person transmission through travel bans or restrictions and screening of travellers
Public health and community level measures such as early detection	Public health and community level measures such as antiviral medication	Public health and community level measures such as surveillance

2. PROCEDURES EMPLOYED TO PREVENT THE SPREAD OF DISEASE

⇒ *2.1 Hygiene practices*

- o Reduce the chance of fomites or food becoming contaminated. And infectious diseases being contracted
- o Washing hands with soap and water
- o Staying home if you are sick
- o Sneezing or coughing into the arm or a tissue
- o Not touching eyes, nose or mouth
- o Not sharing cups, glasses, dishes or cutlery
- o Using separate cleaning cloths for wiping surfaces
- o Proper handling, preparation and cooking of food and proper disposal of contaminated materials to help prevent the spread of disease
- o Clinical effects in hospitals

⇒ *2.2 Quarantine*

- o Limiting or controlling human mobility through travel restriction and quarantine is the first measures applied by national and international public health institutions
- o It includes surveillance, monitoring, screening and clearance procedures at airports and shipping ports

⇒ ***2.3 Vaccination, passive immunity and active immunity***

- o Vaccination helps to prevent diseases in populations and that is the key purpose of immunisation programs
- o They protect an individual against a specific infectious disease and its various complications
- o Vaccines also provide herd immunity for those who cannot be immunised

⇒ ***2.4 Public health campaigns***

- o Focused on prevention and control through strategies such as
 - Health promotions
 - Immunisation campaigns
 - Campaigns to reduce resistance
 - Disease prevention programs

⇒ ***2.5 Use of pesticides***

- o Prevent and control the spread of disease vectors
- o Used in agriculture
- o Can have negative impacts on human health

⇒ ***2.6 Genetic engineering***

- o Can create disease resistant plants
- o Silence genes in targeted pathogens by disabling the gene
- o Engineering the genes of vectors to control the spread of disease
- o Can improve processes for pathogen discovery and diagnosis

3. EFFECTIVENESS OF PHARMACEUTICALS AS TREATMENT STRATEGIES FOR THE CONTROL OF INFECTIOUS DISEASE

⇒ ***3.1 Antivirals***

⇒ ***3.2 Antibiotics***

4. ENVIRONMENTAL MANAGEMENT AND QUARANTINE METHODS TO CONTROL EPIDEMIC AND PANDEMICS

- Aim to control the spread of a disease by reducing exposure to the pathogen through regulation of the environment and modification of behaviours
- Include:
 - Pre-preparedness
 - Alerts
 - Management of healthcare facilities with strict procedures for treatment
 - Use of chemical spray and disinfectants upon entry and exit from facilities
 - The bio secure disposal of all contaminated medical materials
 - Quarantine and isolation of all suspected individuals
 - Exit screening at airports for individuals departing from infected countries

5. DATA RELATED TO INCIDENCE AND PREVALENCE

⇒ *5.1 Mobility of individuals and the portion that are immune or immunised*

- Incidence and prevalence of infectious diseases involves a wide range of interrelated factors including the mobility
- Mobility of individuals in a population influences the incidence and prevalence of disease by changing the frequency of contacts between infected and susceptible individuals
- Mobility also contributes to the incidence and prevalence of infectious diseases and the re-introduction of diseases for the following reasons:
 - Mobility increases contact between humans and vectors
 - New arrivals, refugees, displaced populations and legal and illegal workers bring vectors
 - Changes in global land and animal use
 - Humans encroaching into vector habitats increases the prevalence of diseases for which there was no previous exposure

⇒ *5.2 Malaria or Dengue Fever in South East Asia*

- DENGUE FEVER
 - Factors relating to increases in the incidence and prevalence of dengue fever include
 - Population growth and unprecedented rapid urbanisation resulting in a increased number of people living in poor housing with lack of proper waste which creates favourable breeding conditions for the mosquito vectors
 - Underfunding for prevention and control in many countries
 - Partial or temporary immunity

- Increasing global temperatures
- Increased spread of other vectors other than mosquitos

6. HISTORICAL, CULTURALLY DIVERSE AND CURRENT STRATEGIES TO PREDICT AND CONTROL DISEASE SPREAD

⇒ *6.1 Historical strategies to predict and control the spread of disease*

- Daniel Bernoulli
 - In the 1760s there was a lot of controversy surrounding inoculation as it could protect people but also kill people. In 1760, Bernoulli wrote an article modelling smallpox and the efficacy of vaccination
 - He used Halley's life table and other data to show that inoculation was advantageous. He proved that the risk of dying from inoculation was less than 11%
 - It meant that more people felt comfortable to get inoculated and decreased the spread of smallpox. It increased inoculated rates by 38%

⇒ *6.2 Culturally diverse strategies to predict and control the spread of disease*

- Chinese variolation
 - Aimed to immunize individuals against smallpox
 - Inserting/rubbing powdered smallpox scabs or fluid from pustules into superficial scratches made in the skin. The patient would develop identical pustules caused by the naturally occurring smallpox, usually producing less severe disease
 - It was a risky, but beneficial idea as it worked

⇒ *6.3 Current strategies to predict and control the spread of disease*

- Vaccines
- Campaigns
- Surveillance
- Awareness
- Predictions of future outbreaks

7. COMTEMPORARY APPLICATIONS OF ABORIGINAL PROTOCOLS IN THE DEVELOPMENT OF PARTICULAR MEDICINES AND BIOLOGICAL MATERIALS IN AUSTRALIA AND HOW RECOGNITION OF

INDIGENOUS CULTURAL AND INTELLECTUAL PROPERTY

⇒ *7.1 Aboriginal protocols, medicines and biological materials*

- Much of Indigenous knowledge is crucial to research and development, particularly of pharmaceuticals, agriculture and cosmetic products
- Research into development of traditional medicines and biological materials in Australia requires partnerships with Aboriginal communities
- The Protocol for Aboriginal Knowledge and Intellectual Property was. Developed which set out an ethical guideline the sharing of traditional information

⇒ *7.2 Recognition and protections of Indigenous cultural and intellectual property*

- A major concern for Aboriginals is that their traditional knowledge is being exploited and their Indigenous cultural and intellectual property rights are not recognised
- Practices that do no acknowledge traditional knowledge are not only offensive to many Indigenous people but represent continuous dispossession of their communities

⇒ *7.3 Bush medicine*

- Traditional medicines are relied on by up to 80% of the world's population for primary health care and there is evidence to suggest that ¾ of plants used in pharmaceuticals were originally used in traditional medicine
- Native plants contain antibacterial and anti-inflammatory compounds that are known to Western medicine

⇒ *7.4 Smoke bush in Western Australia*

- In the 1960s, WA government granted the US National Cancer Institute a license to collect plants
- Specimens of this plant were kept until 1981 when they were found to destroy the HIV virus
- The discovery was patented
- Indigenous people are concerned that they have not received any acknowledgement, financial or otherwise for their role in discovering the healing properties of smoke bush

NON INFECTIOUS DISEASES

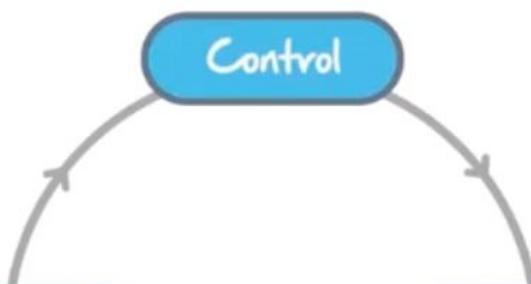
HOMEOSTASIS

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

1. CONSTRUCTING AND INTERPRETING NEGATIVE FEEDBACK LOOPS

- o Homeostasis is the process by which organisms maintain a relatively stable internal environment despite changes in the external environment
- o The body's core temperature, blood pH stays the in range
- o It is when the internal environment contains the right concentration of gases, nutrients, ions and water at the optimal temperature or pH
- o A negative feedback loops is a reaction that causes a decrease in function
- o Occurs in response to some sort of stimulus such as body temperature
- o Causes the output of a system to be lessens so the feedback tends to stabilise the system
- o Without homeostasis chemical reactions cannot act effectively which leads to diseases or medical conditions that negatively impact the organism

Negative feedback loops



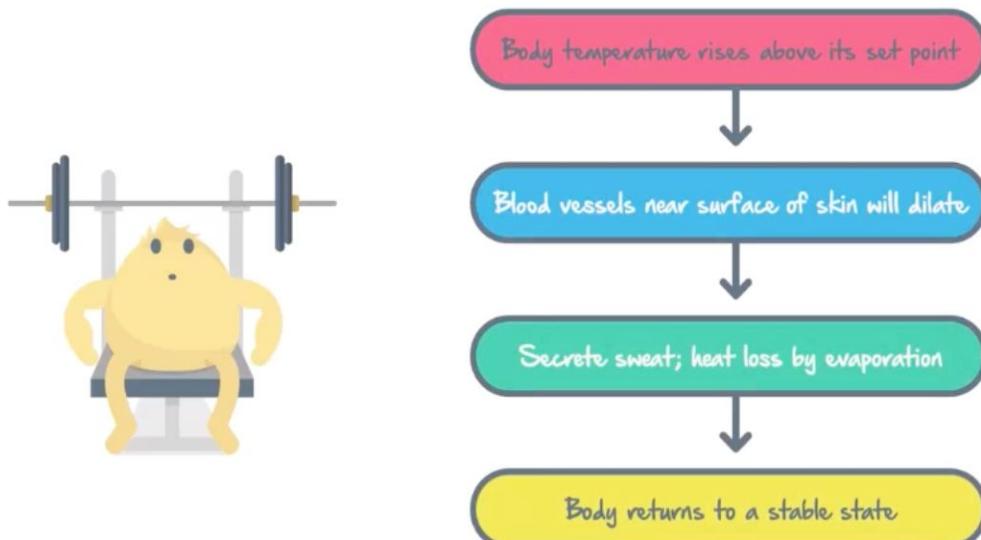
- o The four components of a negative feedback loop are;
 - Stimulus: when the temp. is below or above a set point
 - Receptor: temp. receptors in the skin which communicate info to the brain
 - Control: information is sent to the brain/control centre
 - Effector: blood vessels and sweat glands in the skin react to this information
- o Information is carried from the hypothalamus to the Central Nervous System (CNS)
 - HYPOTHALAMUS: controls certain metabolic processes and other activities of the Autonomic Nervous System. It synthesizes and secretes neurohormones, often called hypothalamic-releasing hormones.
 - CNS: receives the transmitted information, interprets it and sends signals to act upon this information.

⇒ *1.1 Negative feedback loops that maintain homeostasis for temperature*

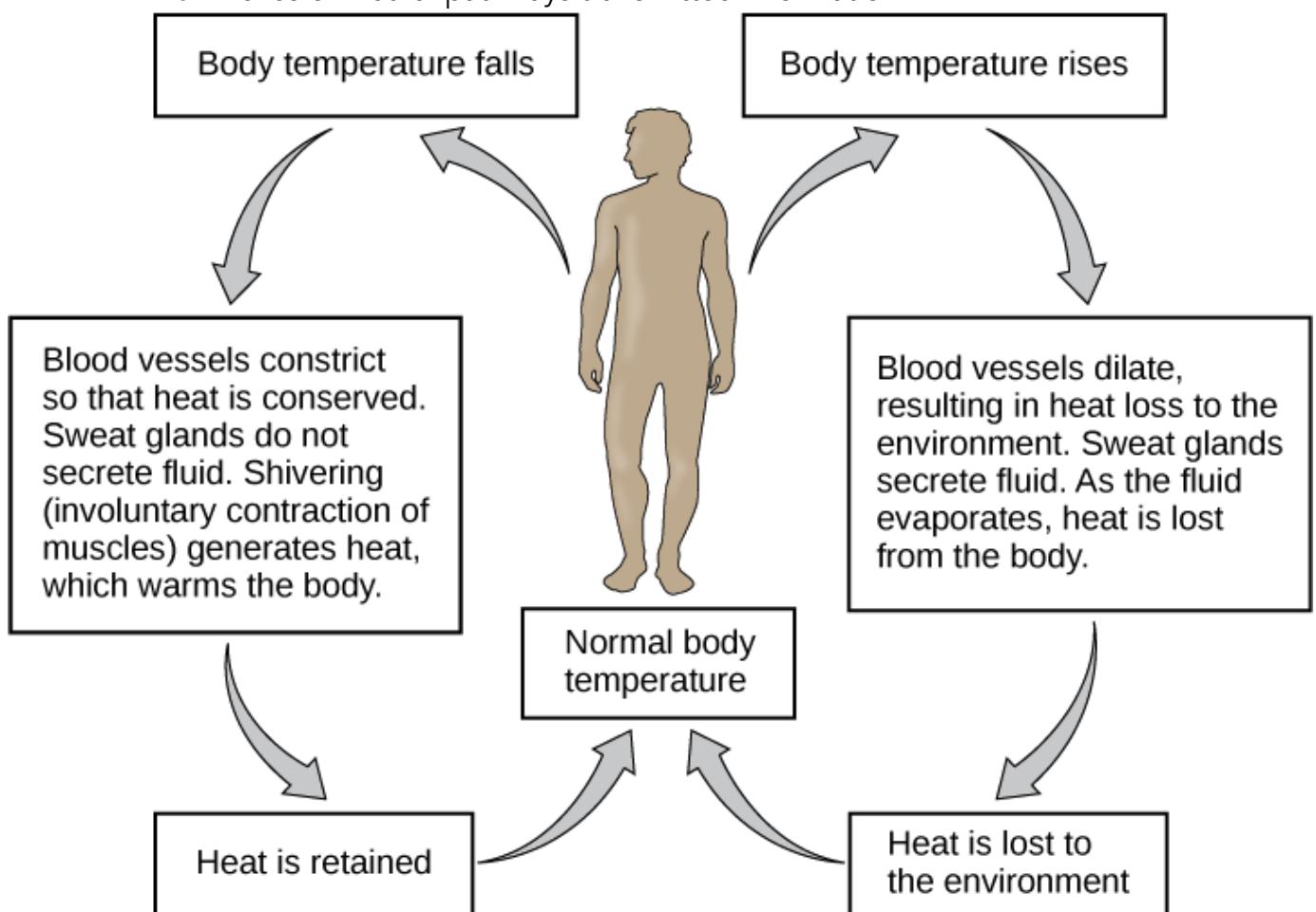
- o It is also called thermo regulation
- o Human internal temperature is between 30-37.5 degrees Celsius
- o When the body is cold;



- When the body is hot;

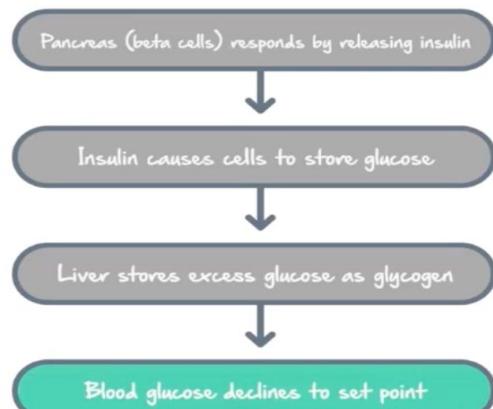
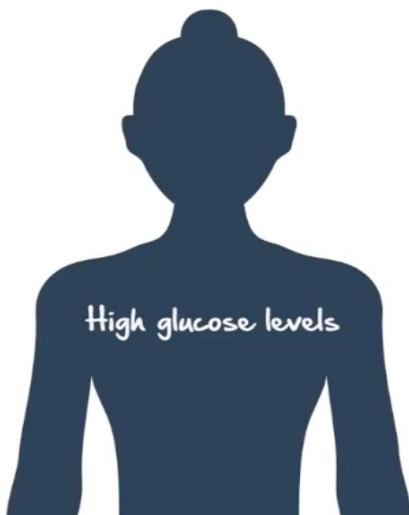


- A constant temp. helps to maintain metabolic processes
- External temp. in mammals is detected by receptors called thermoreceptors
- Internal changes are detected by receptors in the hypothalamus
- Graph looks like an oscillated line
- Relies on neural pathways transmitted information

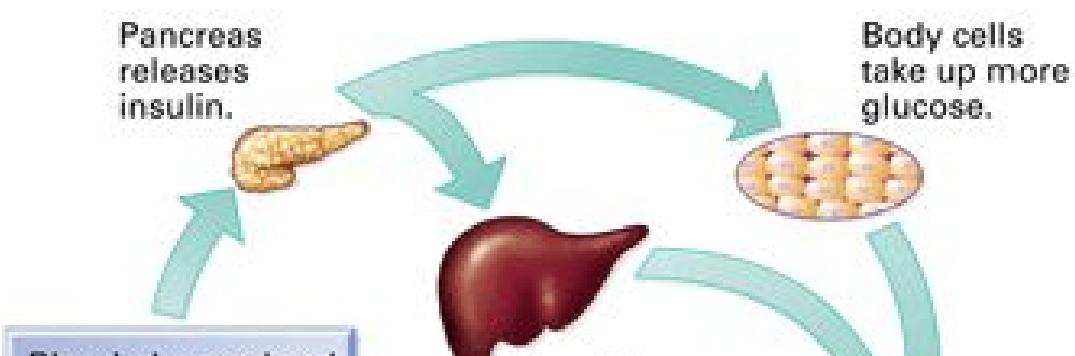
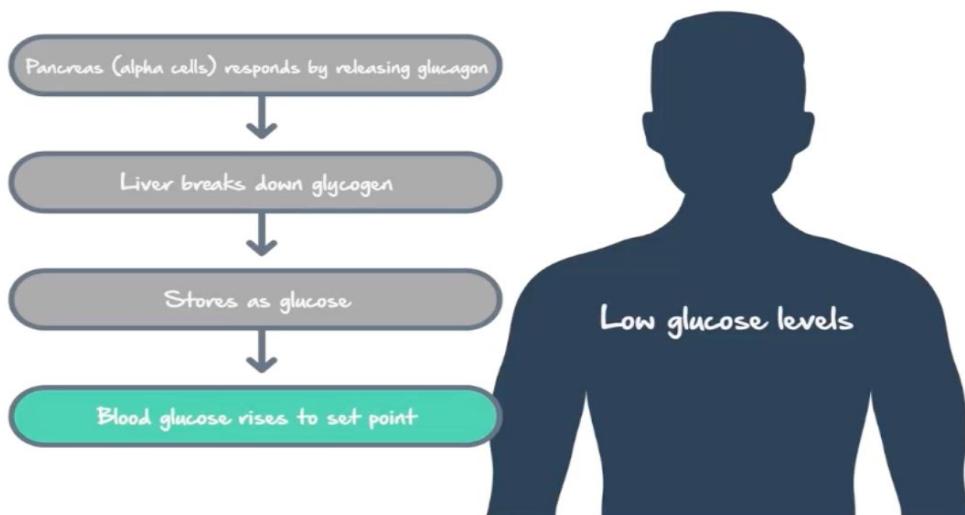


⇒ 1.2 Negative feedback loops to maintain homeostasis for glucose

- It is also chemical regulation
- Chemoreceptor loop
- Relies on hormonal transmitted information
- It is the negative feedback loop of glucose concentration
- When there are high glucose levels;



When there are low glucose levels;



2. MECHANISMS TO MAINTAIN HOMESTASIS

⇒ *2.1 Trends and patterns in behavioural, structural and physiological adaptations in endotherms*

- Endotherms (birds and mammals) generate their own body heat internally and therefore generally maintain a constant body temperature
- Trends and patterns in behavioural, structural and physiological adaptations in endotherms that assist in thermoregulation include;

	Behavioural	Structural	Physiological
Temp. regulation for cooling down or losing heat	Burrowing Nocturnal activity Seeking shade Cooling down in water Stretching out to increase surface area for heat loss	Large ears increase the surface area for evaporative heat loss A larger surface area to volume ratio helps to maximise heat loss	Sweating to increase evaporative heat loss Increased blood flow to the extremities: more blood enters skin capillaries and heat is lost
Temp. regulation for staying warm	Hibernation Migration Curling up in a ball or huddling together to decrease surface area	Insulation such as fat layers, blubber, hair, fur and feathers A smaller surface area to volume ratio helps to minimise heat loss	Reduced blood flow to extremities which keeps the core of the body warm Shivering causes rapid contraction and relaxation of skeletal muscles resulting in more heat being generated Increases in metabolic activity

⇒ *2.2 Internal coordination systems that maintain homeostasis; hormones and neural pathways*

- Nervous system provides rapid and short-term coordination of internal organ systems
- Hormonal system provides slower and longer-lasting response coordination
- NEURAL PATHWAYS
 - Consists of the CNS and peripheral nerves
 - Basic unit is the neuron/nerve cell
 - Nerve cells transmit signals by electrochemical changes in their membranes

- Special endings on the sensory nerves, such as heat sensors or thermoreceptors in the skin detect stimuli such as changes in temperature
- Receptors relay messages that are processed in the central nervous system and then messages are conveyed to effector organs or muscles that bring about the response
- o HORMONES
 - Produced by the endocrine system
 - They alter the metabolism of target cells, tissues or organs by increasing or decreasing their activity
 - If a person begins to dehydrate, their blood volume drops
 - Change is detected by hypothalamus which stimulates pituitary to secrete ADH hormone which stimulates nephrons in the kidney to reabsorb more water. This decreases urine production, increases urine concentration and maintains the concentration of water in the blood.

⇒ *2.3 Mechanisms in plants that allow water balance to be maintained*

- o Plants have specific adaptations for obtaining water and minimising water loss as well as metabolic processes
- o Australian sclerophyll plants have a range of adaptations to minimise water loss
 - Thin leaves with a waxy cuticle
 - Sunken stomata on the underside
 - Leaves hung vertically to reduce surface area exposed to the sun

CAUSE AND EFFECT

INQUIRY QUESTION: Do non-infectious diseases cause more deaths than infectious diseases?

1. INVESTIGATING THE CAUSES AND EFFECTS OF NON-INFECTION DISEASES IN HUMANS

⇒ *1.1 Genetic diseases*

- o Inherited disorders that are caused by genetic mutations passed down from parents to offspring
- o Mutations in germ-line cells result in a large proportion of inherited disorders

Examples	Cause of disease	Effects
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Sickle-cell anaemia	Caused by a mutation in a gene that tells the body to make haemoglobin; blood cell becomes misshapen and breaks down; recessive disorder	Not enough healthy red blood cells to carry enough oxygen which leads to fatigue, pain and frequent infections
Down syndrome	Caused by an additional chromosome 21	Mild to moderate mental retardation and poor muscle tone
Haemophilia	Sex-linked genetic disorder; the gene that controls blood clotting proteins	Males with haemophilia do not have the genetic information for blood clotting
Huntington's disease	Toxic protein that collects in the brain and causes damage to brain and nerve cells	Progressive brain disorder that causes uncontrolled movements, emotional problems and loss of cognitive function

⇒ 1.2 Diseases caused by environmental exposure

- Caused by exposure to the environment
- Includes stress, noise, overcrowding, drugs and pollutants
- Mutagens can be classed as physical or chemical and this often causes non-infectious diseases

Examples	Cause of disease	Effects
Mesothelioma	Rare type of cancer effecting the membrane that protects the lungs	Tiny fibres of asbestos are breathed in or swallowed which causes inflammation
Skin cancer	Excessive exposure to UV radiation Causes melanoma that develops when a mole becomes cancerous	Most lethal form of skin cancer and tumours start in the pigment-producing melanocytes in the skin
Asthma	Inflammatory disease of the airways though the causes are not fully understood	Vary in severity but include breathlessness, wheezing, tight feeling, coughing

⇒ 1.3 Nutritional diseases

- Caused by a nutritional deficiency of an important compound such as a protein, vitamin or mineral
- Can also include diets, unhealthy eating, alcohol consumption, poor food choices, tobacco smoking and lack of physical exercise
- Eating disorders are also classed as nutritional diseases

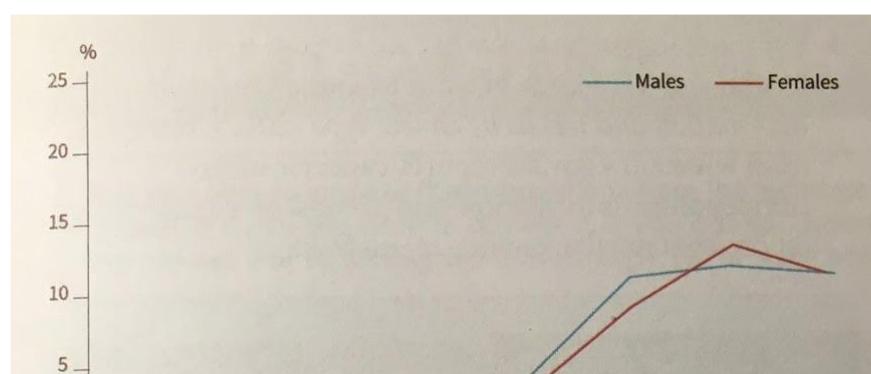
Example	Cause of the disease	Effects
Vitamin k deficiency	Lack of green leafy vegetables. In the diet	Prevents blood from clotting, leading to serious blood loss
Anaemia	Iron deficiency	Reduces the production of healthy blood cells so the blood is unable to carry enough oxygen to the tissues
Night blindness	Vitamin A deficiency	Vision problems as vitamin A is critical for the formation of light receptors
Kwashiorkor	Severe form of malnutrition caused by a protein deficiency	Emaciated body except for the ankles, feet and belly which swell with fluid

⇒ 1.4 Cancer

- Cancer is a disease of the body's cells where a mutation occurs in the cells' DNA and they multiply in an uncontrollable way
- Cancer/tumour describes the collections of these cells growing and potentially spreading within the body. Cancerous cells can arise from almost any type of tissue
- Tumours are either benign or malignant
- Mutagens that cause cancers are referred to as carcinogens

Examples	Cause of the disease	Effects
Breast cancer	Mutations in genes due to carcinogen exposure leading to DNA damage	Breast cancer Cancer of the breasts
Blood cancers	Smoking, exposure to petrochemicals, chemotherapy and a family history	Blood cancer Cancer of the blood
Lung cancers	Tobacco smoking Carcinogens damage the cells that line the lungs	Can't breathe Lungs are unable to function

2. DATA THAT



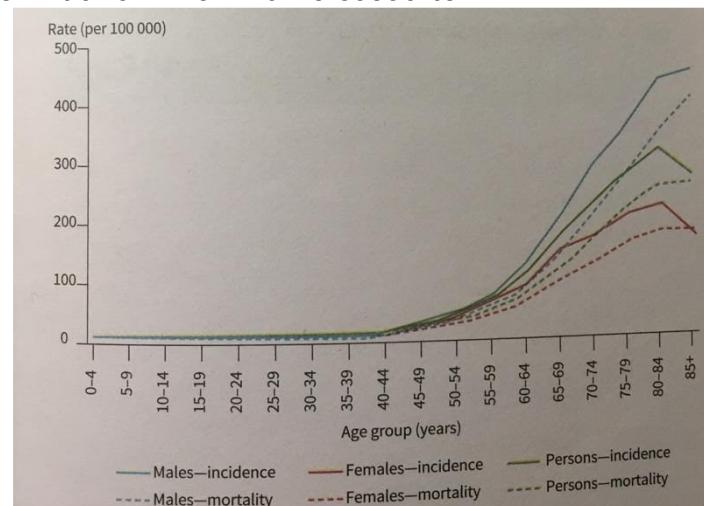
SHOWS THE INCIDENCE, PREVALENCE AND MORTALITY RATES OF NON-INFECTIOUS DISEASE

⇒ 2.1 Nutritional diseases

- Diabetes data:
 - 2015 there were 16400 deaths due to diabetes with 55% being type 2
 - In 2016 diabetes was Australia's 7th leading cause of death
 - ABS 1.2 million people had diabetes and 85% had type 2

⇒ 2.2 Diseases caused by environmental exposure

- Lung cancer data:
 - Is the leading cause of death and was the fifth most common cancer in Australia 2015
 - 2014, 8251 people died from it and in 2017 it increased to 9021
- Melanoma skin cancer
 - Fourth most commonly diagnosed cancer in Australia 2013
 - 10th leading cause of cancer deaths in Australia
- Prevalence is the number or proportion of cases in a population at a given time
- Incidence is the number of new cases diagnosed in a given time period
- Mortality refers to the number of deaths in a specified time period



EPIDEMIOLOGY

INQUIRY QUESTION: Why are epidemiological studies used?

1. ANALYSING PATTERNS OF NON-INFECTIOUS DISEASES IN POPULATIONS

⇒ 1.1 Nutritional diseases

- TYPE 2 DIABETES
 - Australians vs. Indigenous Australians:
 - IA = 3.5x more likely to have diabetes

- IA = 4x more likely to be hospitalised
- Major cities vs. remote cities
 - RC = 1.8x more likely to be hospitalised
 - RC = 1.9x more likely to die from it
- High socio-economic vs. low socio-economic
 - LSE = 3.6x more likely to have diabetes

⇒ ***1.2 Diseases caused by environmental exposure***

- MELANOMA CANCER
 - Survival improved from 86% - 90%
 - Number of new cases increased

2. INVESTIGATE THE TREATMENT/MANAGEMENT AND POSSIBLE FUTURE DIRECTIONS FOR FURTHER RESEARCH, OF A NON-INFECTIOUS DISEASE USING AN EXAMPLE

⇒ ***2.1 Treatment and management of non-infectious diseases***

- Includes primary health care, specialised treatment services and targeted services for populations most at risk
- Fundamental aim is to prevent disease so that people remain as healthy as possible for as long as possible
- Survival rates for non-infectious diseases have improved significantly in Australia since 1980
- TREATMENT AND MANAGEMENT OF MELANOMA
 - Depends on the stage of the disease, location and severity of symptoms
 - Surgery is the main initial treatment
 - Thin early stage melanomas can be removed by minor surgery
 - A wide margin of healthy skin around the edge of the tumour is removed to ensure that no cancer cells are left behind
 - The thicker the tumour, the wider the margin required in terms of both area and depth
 - If the melanoma has spread to nearby lymph nodes these lymph nodes are usually removed by surgery
 - Surgery is not necessarily a cure but it might prolong survival and avoid the pain
 - Additional treatments include:
 - Chemotherapy: which is less effective for melanoma but may be used to prolong survival or relieve symptoms
 - Radiotherapy: not usually used to treat the original melanoma in the skin but may be used to treat melanoma that has recurred, to kill any cancer cells remaining after surgery or to relieve symptoms caused by spread of melanoma

- Immunotherapy: uses medicines to block proteins on immune system cells that stop these cells from attacking other cells in the body

⇒ *2.2 Possible future directions*

- Cancer vaccines
- Discovery of proteins in T Cells
- Proton therapy
- Focused radiotherapy
- Monoclonal antibody that targets antigens
- Novel technologies for early detection

3. EVALUATE THE METHOD USED IN AN EXAMPLE OF EPIDEMIOLOGICAL STUDY

4. EVALUATE THE BENEFITS OF ENGAGING IN AN EPIDEMIOLOGICAL STUDY

- Uncover the changes in prevalence and incidence data over time and/or differences in prevalence among populations help to identify risk factors, determine the resources needed for health care, education and research and develop targeted public health interventions that are cost and resource effective

P R E V E N T I O N

INQUIRY QUESTION: How can non-infectious diseases can be prevented?

1. EFFECTIVENESS OF CURRENT DISEASE PREVENTION METHODS AND STRATEGIES

⇒ *1.1 Educational programs and campaigns*

- Growing body of evidence in Australia about the prevention methods and strategies that work
- Include taxation, media campaigns and regulation of unhealthy products

EDUCATION PROGRAM/CAMPAIGN	TARGET

Crunch and Sip	School-based programs targeting diet and nutrition to prevent diseases such as overweight, obesity and type 2 diabetes in children and young people
Jump Rope for Heart	School-based program aimed at keeping kids healthy and raising funds to fight heart disease
Swap It, Don't Stop It	Community-wide campaign to encourage people to make small lifestyle changes for big improvements in health
Slip, Slop, Slap, Seek, Slide	Community-wide sun protection education program to encourage people to slip on a shirt, slop on sunscreen, slap on a hat, seek shade and slide on sunglasses
Sun Smart	Community-based, state-wide media campaigns to change broader social norms about sun protection
Quit Smoking and I Can Quit	Anti-smoking education and mass media campaigns target specific groups such as young people or Indigenous people
Don't Turn a Night Out into a Nightmare	National anti-binge drinking campaign targeting teenagers and young adults

⇒ **1.2 Genetic engineering**

- Association of suspected carcinogenic exposure and cancer risk in populations is studies
- Find which SNPs are associated with a disease
- Find the probability of non-infectious diseases being passed on to offspring

TECHNOLOGIES AND DISORDERS

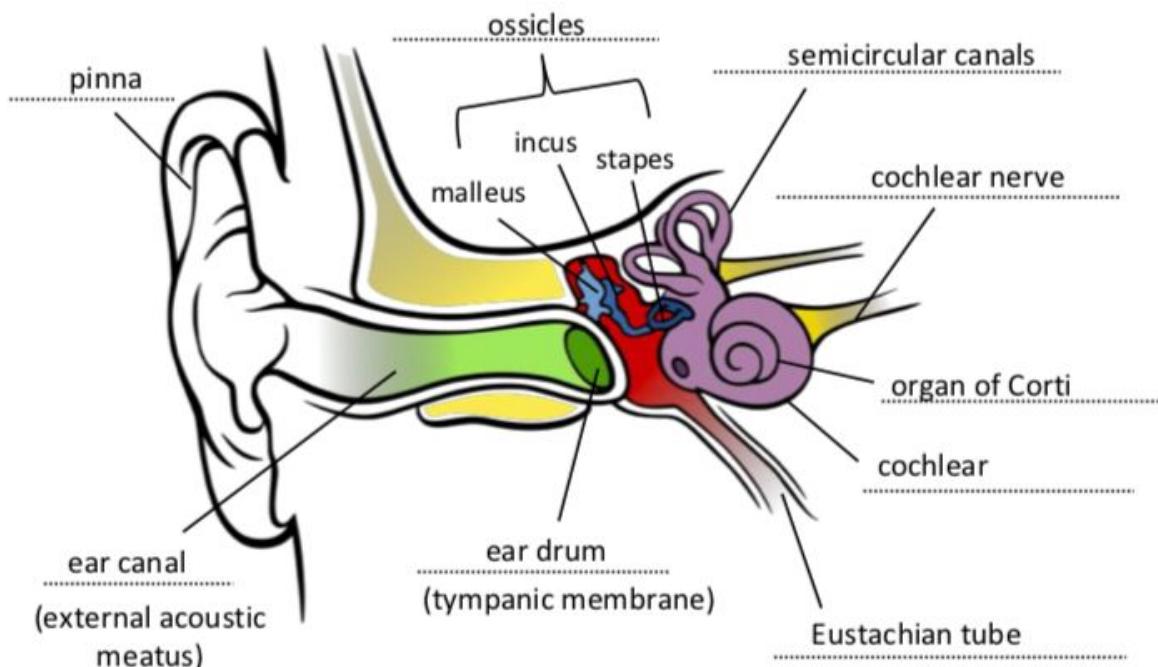
INQUIRY QUESTION: How can technologies be used to assist people who experience disorders?

1. EXPLAIN A RANGE OF CAUSES OF DISORDERS AND INVESTIGATE THE STRUCTURE AND FUNCTION OF ORGANS

⇒ 1.1 Hearing loss

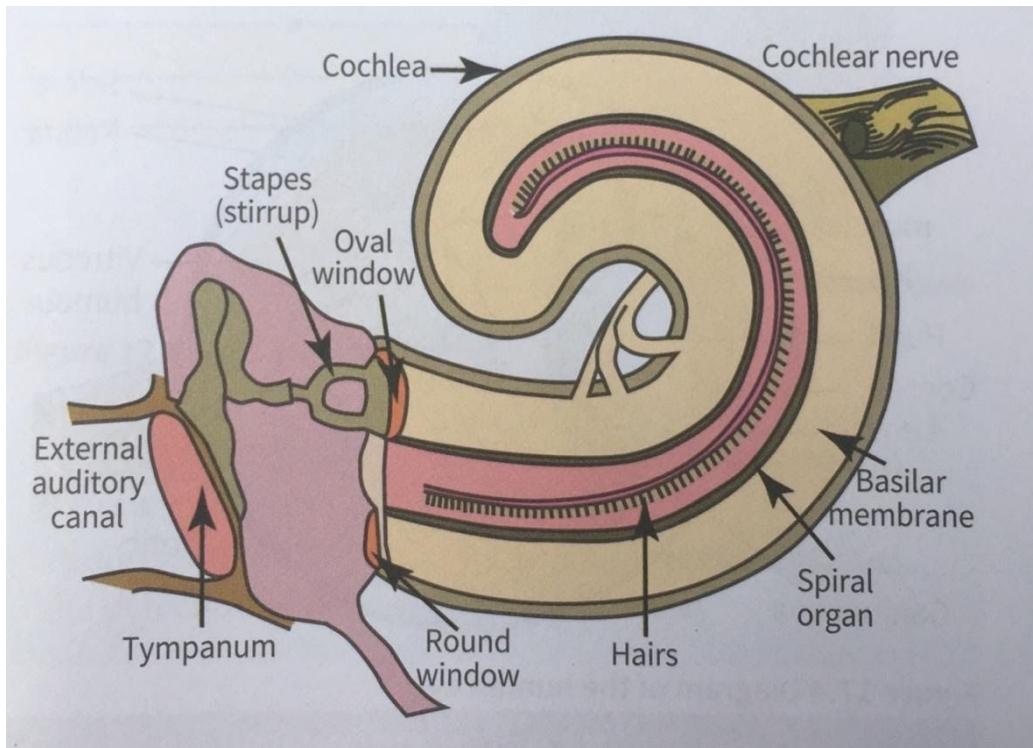
- THE STRUCTURE AND FUNCTION OF THE EARS

- Ears provide hearing and balance
- Ears and brain interpret changes in frequency, tone, volume or sounds in the environment
- Humans can't hear sounds > 20,000hz
- Sounds with short wave lengths have high frequencies
- Sounds with long wave lengths have low frequencies
- Sounds vary in amplitude and loudness
- The loudness of the sound is determined by the energy carried by the wave



STRUCTURE	FUNCTION
PINNA (THE FLAP)	<ul style="list-style-type: none"> - Collects sound waves from a wide area - Funnels the sound into the external ear passage
TYMPANIC MEMBRANE (EARDRUM)	<ul style="list-style-type: none"> - Stretched across the end of ear canal - Separates the outer ear from the middle ear - Sound waves cause the ear drum to vibrate - This is conveyed from the eardrum to the oval window by the ossicles
EAR OSSICLES	<ul style="list-style-type: none"> - Three bones; hammer, anvil and stirrup - Transmit the sound waves to the inner ear - Sound vibrations travel well through bone
OVAL WINDOW AND ROUND WINDOW	<ul style="list-style-type: none"> - Oval window and round window are two thin membranes

	<ul style="list-style-type: none"> - Sound reaches the inner ear at the oval window and pressure is then transmitted in the fluid in the tympanic canal - Pressure causes the round window at the tympanic canal, to bulge outwards - The oval window helps to amplify the pressure of the sound waves
COCHLEA (SNAIL-LIKE SPIRAL COILED TUBE IN THE INNER EAR)	<ul style="list-style-type: none"> - Cochlear contains the receptors for sound and the vestibular apparatus that is associated with a sense of balance - As a result, the round window bulging outwards, fluid in the cochlear tubes vibrates
ORGAN OF CORTI	<ul style="list-style-type: none"> - Contains the auditory cells
AUDITORY NERVE	<ul style="list-style-type: none"> - Transmits the sound vibrations to the brain



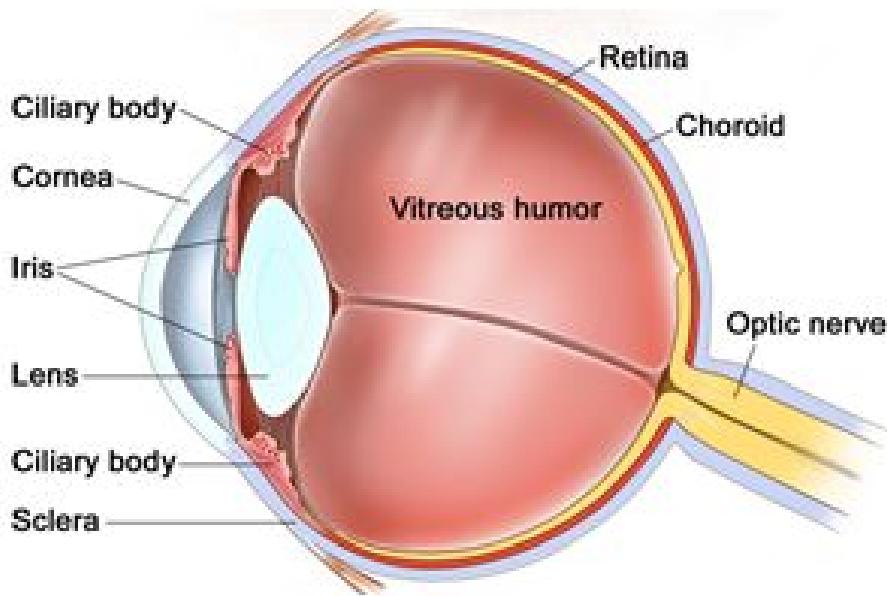
- Sound waves must travel through a medium, which they do by vibrating the molecules in the medium
- Sound waves travel through the ear canal and cause the ear drum to vibrate
- These vibrations are transmitted to the hammer, anvil and stirrup of the middle and inner ear
- Sounds reaches the inner ear at the oval window
- Vibrations are transmitted to the spiral cochlea which contains fluid and nerve endings
- Vibration at the inner ear, displacement of cochlear fluid and movement of hair cells of the organ of Corti produce electrochemical signals

- Auditory nerve transmits these neural impulses to the auditory area of the cerebrum where they are interpreted
- o CAUSES OF DISORDERS IN THE STRUCTURES AND FUNCTIONS OF THE EARS: HEARING LOSS
 - Good hearing requires good conduction of the sound waves through the outer ear, proper mechanical working of the ear ossicles, healthy nerve pathways for the signals to the brain
 - If anything goes wrong with the structures and their functions a person will have some form of hearing loss
 - An individual's hearing loss varies depending on the pitch of the sounds they can't hear
 - Can be different in each ear
 - Hearing loss is measured in decibels

TYPE OF HEARING LOSS	CAUSES
Conductive hearing loss due to problems with the ear canal, eardrum, middle ear or ear ossicles	<ul style="list-style-type: none"> - Blocked ear canal caused by excessive earwax which decreases the volume of the sound being transmitted - Infection in the ear canal - Perforated eardrum caused by loud noise (explosion), poking the inner ear, injury from a severe blow
Sensorineural hearing loss due to problems with the inner ear	<ul style="list-style-type: none"> - Damage to the hair or nerve cells in the cochlea caused by aging or cumulative exposure to excessive noise or certain ototoxic drugs - When hairs are damaged, electrical signals are not transmitted efficiently - Large portions of the hair cells of the organ of Corti are damaged or missing - Head trauma - Infection from a virus such as measles, mumps or meningitis - Malformation of the inner ear - Autoimmune inner ear disease
Mixed hearing loss	<ul style="list-style-type: none"> - Combination of conductive and sensorineural hearing loss to the outer, middle and inner ear or auditory nerve; therefore, a possible combination of causes

⇒ 1.2 Visual disorders

- THE STRUCTURE AND FUNCTION OF THE EYES
 - The eye is responsible for both vision and balance
 - The structure of the eye relates to its function of admitting light, refracting and focusing light to form an image and converting that image into nerve impulses that are then conveyed to the brain
 - Light does not need a medium to travel through
 - Light can pass through objects, be reflected or refracted



STRUCTURE	FUNCTION
CONJUNCTIVA	Is a thin protective covering of epithelial cells. It protects the cornea against damage by friction (tears help this process by lubricating the surface of the conjunctiva)
CORNEA	Is the transparent, curved front of the eye which helps to converge the light rays which enter the eye
SCLERA	Is an opaque, fibrous, protective outer structure. It is soft connective tissue, and the spherical shape of the eye is maintained by the pressure of the liquid inside. It provides attachment surfaces for eye muscles
CHOROID	Has a network of blood vessels to supply nutrients to the cells and remove waste products. It is pigmented that makes the retina appear black, thus

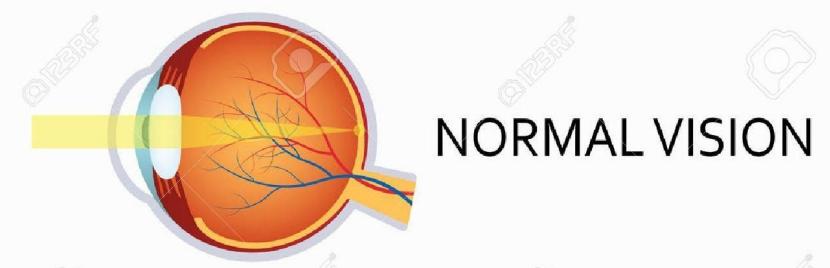
	preventing reflection of light within the eyeball
CILIARY BODY	Has suspensory ligaments that hold the lens in place. It secretes the aqueous humour, and contains ciliary muscles that enable the lens to change shape, during accommodation (focusing on near and distant objects)
IRIS	Is a pigmented muscular structure consisting of an inner ring of circular muscle and an outer layer of radial muscle. Its function is to help control the amount of light entering the eye to that: <ul style="list-style-type: none"> - Too much light does not enter the eye which would damage the retina - Enough light enters to allow a person to see
PUPIL	Is a hole in the middle of the iris where light is allowed to continue its passage. In bright light it is constricted and in dim light it is dilated
LENS	Is a transparent, flexible, curved structure. Its function is to focus incoming light rays onto the retina using its refractive properties
RETINA	Is a layer of sensory neurones, the key structures being photoreceptors (rod and cone cells) which respond to light. Contains relay neurones and sensory neurones that pass impulses along the optic nerve to the part of the brain that controls vision
FOVEA	A part of the retina that is directly opposite the pupil and contains only cone cells. It is responsible for good visual acuity (good resolution)
BLIND SPOT	Is where the bundle of sensory fibres forms the optic nerve; it contains no light-sensitive receptors
VITREOUS HUMOUR	Is a transparent, jelly-like mass located behind the lens. It acts as a 'suspension' for the lens so that the delicate lens is not damaged. It helps to maintain the shape of the posterior chamber of the eyeball

AQUEOUS HUMOUR

Helps to maintain the shape of the anterior chamber of the eyeball

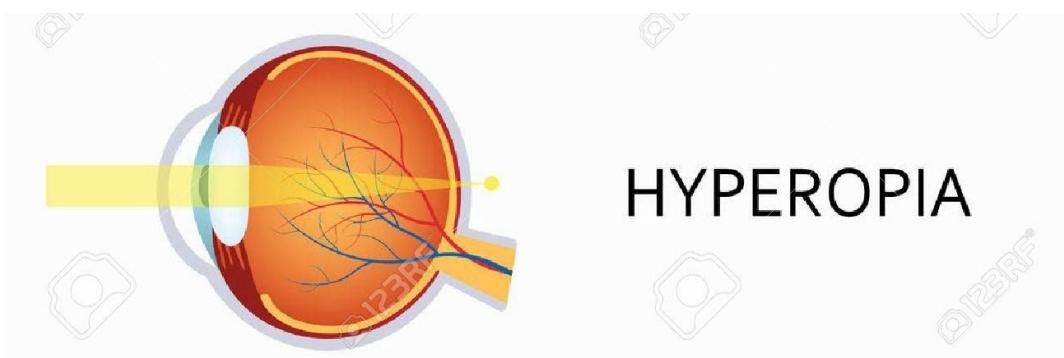
o CAUSES OF DISORDERS IN THE STRUCTURES AND FUNCTIONS OF THE EYES: VISUAL DISORDERS

- Good eyesight requires good transmission of light, correct refraction of light, precise focal length for focusing the light and functional nerve pathways that take the signal to the brain
- When things go wrong, a person has visual disorders/blind



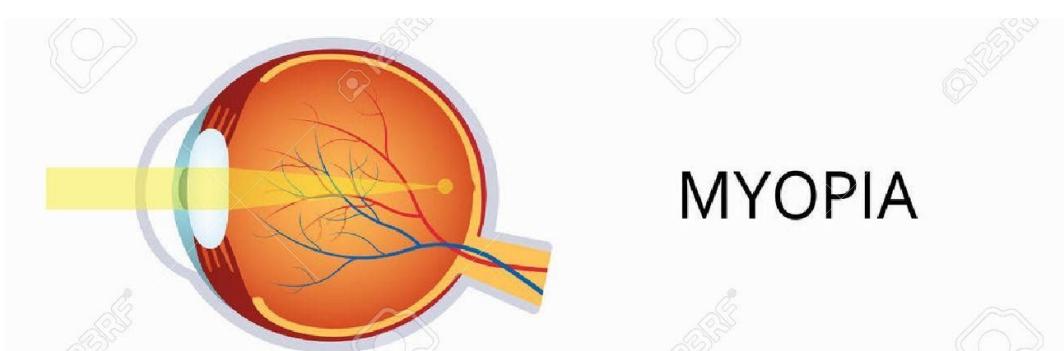
NORMAL VISION

-
-
- Hyperopia: is called long-sightedness
 - Happens if the cornea is flatter or the eyeball is shorter, or the lens is positioned further back than normal
 - Near objects are not in focus



HYPEROPIA

- Myopia: is called near, or short-sightedness
 - Happens if the cornea is more curved, the eyeball is longer, or the lens is positioned further forward than normal. The image produced is in front of the retina
 - Distant objects are not in focus



MYOPIA

- Hyperopia and myopia are caused by genetic and environmental factors
- Both tend to run in families although close-up work such as reading, and eye strain can increase the chances of myopia

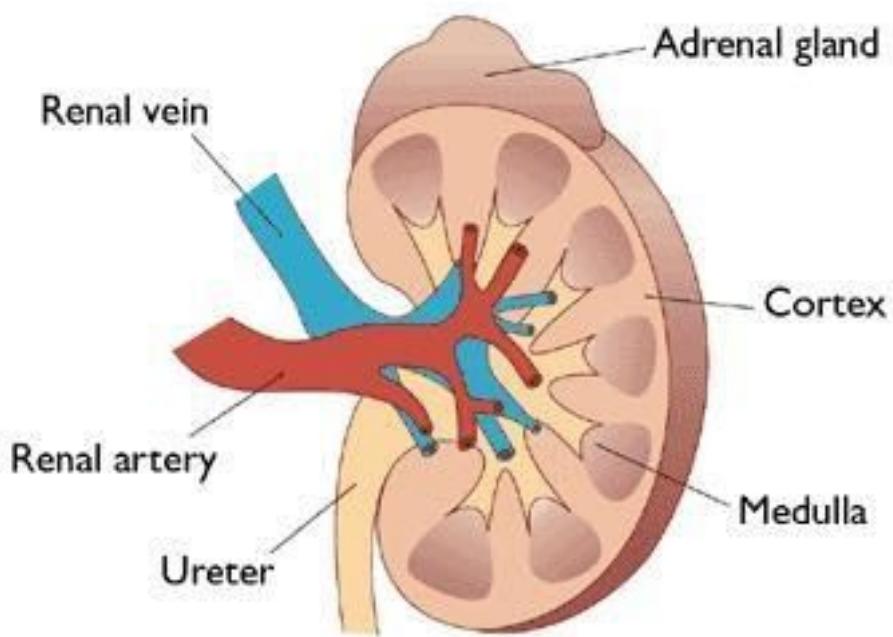
TYPE OF VISUAL DISORDER	CAUSE
Astigmatism	A refractive error in which the eye does not focus the light evenly on the retina
Cataracts	A degenerative condition in which lens of the eye clouds over, obstructing the passage of light to cause vision loss and potentially blindness; it is the result of deterioration of proteins in the lens
Macular degeneration	A progressive deterioration of the macula of the retina (the central inner-lining); it is often related to old age and results in a loss of central vision
Diabetic retinopathy	A complication of diabetes caused by prolonged high blood glucose levels; over time high blood glucose levels weaken and damage blood vessels within the retina, starving the retina of oxygen and causing haemorrhage or swelling Diabetic retinopathy occurs in 15% of Australians with type 1 diabetes
Glaucoma	Caused by damage to the optic nerve due to increased pressure within the eyes
Night blindness	Caused by short-sightedness, cataracts or vitamin A deficiency
Childhood blindness	Caused by a number of factors such as abnormalities in the eye itself and congenital blindness as a result of damage to the brain due to lack of oxygen at birth
Colour blindness	A genetic disorder caused by the lack of one or more of the colour-sensitive pigments in the cones the most common type of colour blindness is red-green in which the cones most receptive to red light and green light are missing

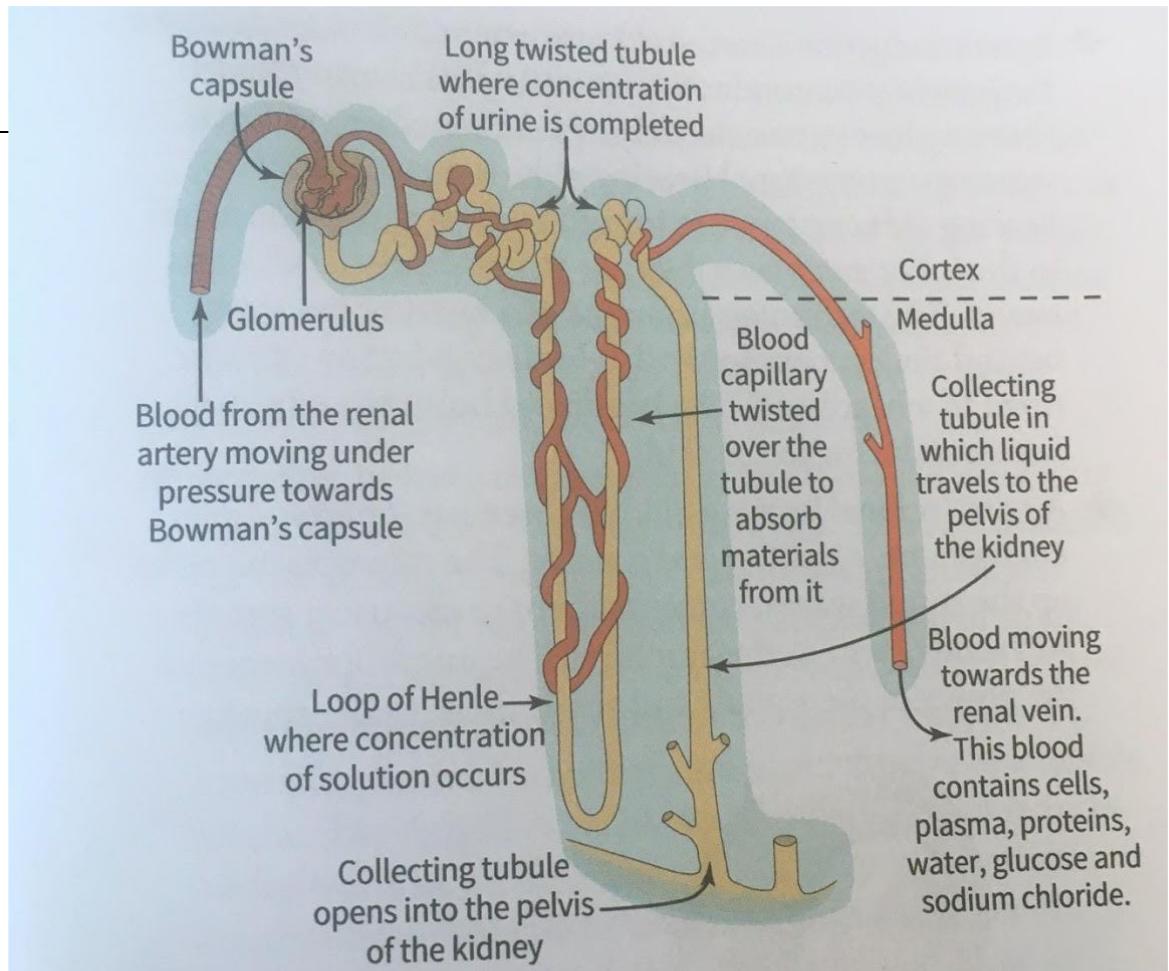
⇒ 1.3 Loss of kidney function

- o STRUCTURE AND FUNCTION OF KIDNEYS
 - Kidneys are complex organs of the excretory system

- The main functions of the kidneys are to filter wastes from the blood and produce urine without the loss of too much water
- The main wastes are nitrogenous products
- These wastes are produced when proteins are metabolised
- They are toxic and must therefore be removed from the body
- Kidneys help in the homeostasis of water, glucose and inorganic salts
- The composition, quality and quantity of urine produced depends on factors such as a diet, physical exercise, metabolism and water intake
- The renal artery brings blood carrying wastes and other substances to the kidneys
- Urine is formed in the cortex and medulla, collected in the pelvis and delivered to the bladder through the ureter
- The renal vein returns filtered blood to the body
- In the human kidney water reabsorption is a passive process of diffusion and osmosis
- Reabsorption of sodium salts, glucose and amino acids are active processes that require energy
- The functional unit of the kidney is the nephron
- Nephrons are arranged in the outer cortex and central medulla
- Each nephron is made up of:
 - A filtering unit
 - Areas where both passive and active reabsorption take place
 - A collecting area for urine

Diagram of Kidney





STRUCTURE	FUNCTION
BOWMAN'S CAPSULE	A cuplike sac that encloses a glomerulus; it functions to carry out the first step in filtration of blood from the urine
GLOMERULUS	A ball of capillaries within the Bowman's capsule that acts as an 'ultra' filter
LONG TWISTED TUBULES AND LOOP OF HENLE	The twisted tubules occur in the cortex; the Loop of Henle is the u-shaped part of the nephron and it extends from the cortex down into the medulla and back; the tubules play a role in the reabsorption of substances
COLLECTING TUBE	Long straight tubule which opens into the pelvis of the kidney; the wall of the collecting tubule may be permeable or impermeable to water

- If the all of the collecting tube is permeable to water, water is reabsorbed into the surrounding tissues
- Concentrated urine is excreted, and water is conserved
- If the wall of the collecting tubule is impermeable to water, no water is reabsorbed, and dilute urine is excreted

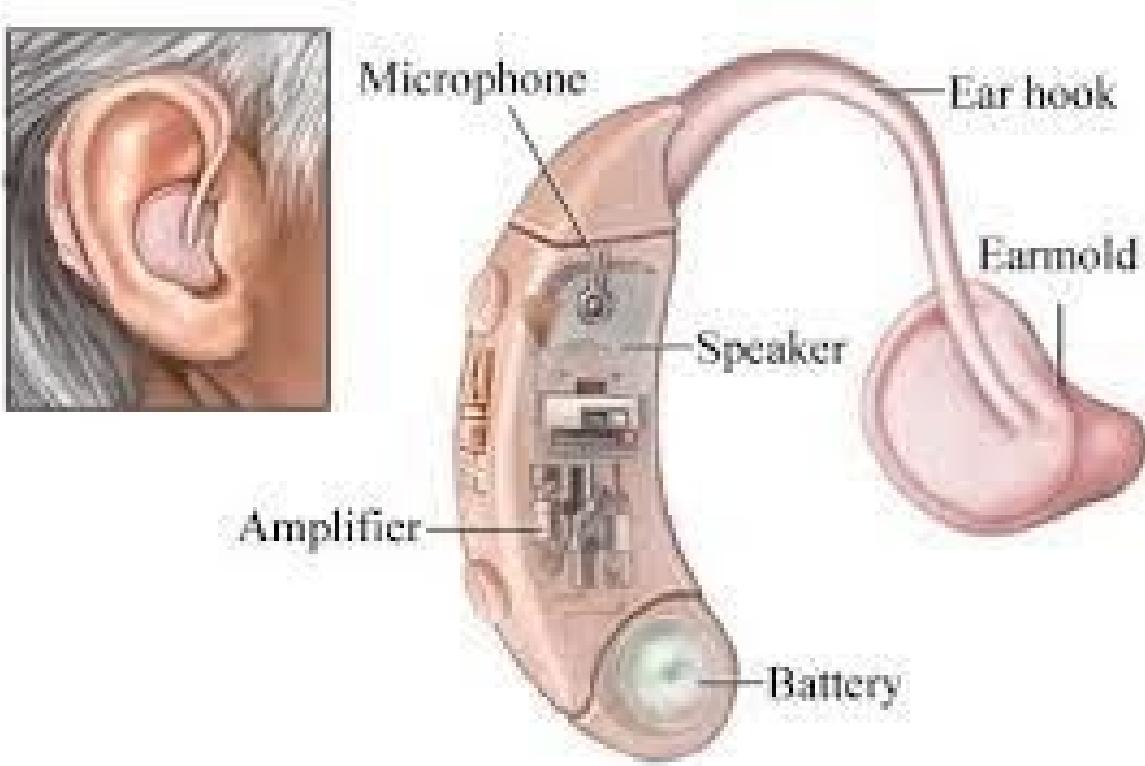
- The permeability of the wall is controlled by the antidiuretic hormone (ADH)
- The adrenal cortex with the adrenal glands produces two hormones: cortisol and aldosterone
- Cortisol controls many metabolic processes including blood pressure
- Aldosterone functions to maintain the balance of water and salts in the body
- The hormone stimulates the nephrons to decrease reabsorption of potassium and increase reabsorption of sodium into the blood
- o CAUSES OF DISORDERS IN THE STRUCTURE AND FUNCTIONS OF THE KIDNEYS: LOSS OF KIDNEY FUNCTION
 - A healthy functioning kidney needs structures and processes for filtration, active and passive reabsorption of substances, and diffusion and osmosis
 - When things go wrong with these structures and processes a person has loss of kidney function
 - Disorders of the kidneys include kidney disease and kidney failure
 - Kidney disease occurs when the nephrons are damaged
 - Kidney disease is often called a 'silent disease' as up to 90% of kidney function can be lost before symptoms appear
 - As a result, many people are unaware they have a condition
 - Chronic kidney disease is caused by risk factors such as smoking, high blood pressure, overweight and obesity, and diabetes
 - The most severe form of chronic kidney disease is end-stage kidney disease where people usually require kidney replacement therapy, or dialysis to survive
 - Acute kidney injury occurs when an abrupt loss of kidney function causes the body to accumulate waste products and become unable to maintain homeostasis
 - Other diseases include:
 - Nephritic disease
 - Nephrosis
 - Kidney stones

2. INVESTIGATES TECHNOLOGIES THAT ARE USED TO ASSIST WITH THE EFFECTS OF A DISORDER

⇒ *2.1 Hearing loss: cochlear implants, bone conduction implants, hearing aids*

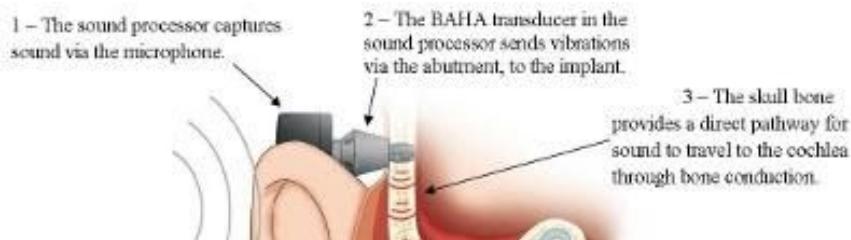
- o HEARING AIDS
 - Electronic devices that can amplify sounds entering the external ear
 - Worn behind the ear or in the ear canal

- They have a microphone to detect and convert sound into electrical signals
- They have an amplifier to strengthen these signals
- They have a receiver to convert the signals back to sound
- They have 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 larger vibrations
- Today, there are many different hearing aids available
- Many can be programmed to dull background noise and to selectively amplify frequencies for which hearing loss has occurred



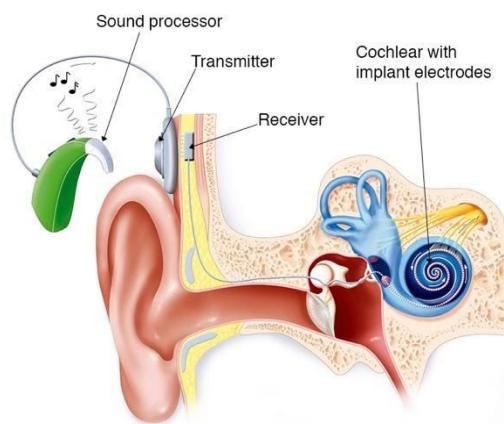
o BONE CONDUCTION IMPLANTS

- 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
- External sound processor is attached either magnetically or directly to a small titanium implant that is surgically placed in the bone behind the ear
- Sound processor detects and converts sound vibrations
- These are transferred directly through the bone to the cochlea
- Hair cells in 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 conduction treatment prior to surgery



- o COCHLEAR IMPLANTS

- Also known as bionic ears
- They are used to treat sever-to-profound hearing loss with missing or damaged hair cells in the cochlea
- It is worn externally behind the ear as a sound processor that detects sound and converts it into digital code
- Digital code is transmitted to an implant under the skin
- The implant converts the digital code into electrical impulses and sends them along electrodes into the cochlea
- The impulses directly stimulate the cochlear nerve
- The signals are recognised by the brain as sound, allowing a person to hear
- Cochlear implants work best when implanted before the age of 5
- They do not fully reproduce the sounds experienced by someone with full hearing and do not work if the auditory nerve is damaged
- Once fitted, the patient has to be trained to interpret the sounds they 'hear' for the first time
- Cochlear implants are very expensive due to production costs, the need to surgically implant the electrodes and ongoing costs to maintain the external parts
- However, for people with significant hearing loss, cochlear implants facilitate hearing
- This is particularly helpful in allowing young children with hearing loss to learn spoken language

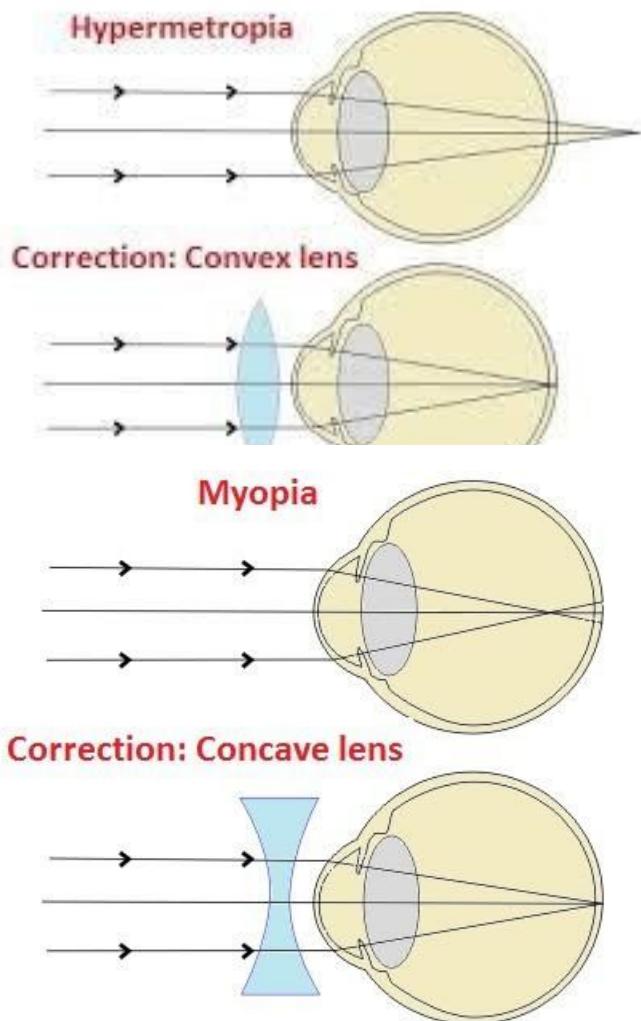


⇒ **2.2 Visual disorders: spectacles, laser surgery**

- o SPECTACLES

- Whether visual disorders are treatable depends on the cause

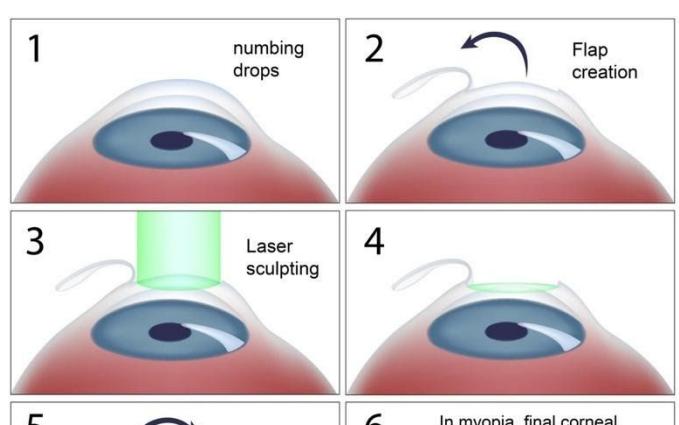
- Technologies that assist with the effects of visual disorders include spectacles
- Prescription spectacles and contact lenses are artificial lenses designed to correct the refractive errors of myopia and hyperopia



o LASER SURGERY

- Form of vision correction that involves reshaping the cornea to correct refractive error
- The technology uses cool temperature laser
- LASIK surgery is the most common form of laser surgery
- It involves two steps as outlined below
 - A thin flap is created in the cornea to allow it to be reshaped; the laser then removes the exact amount of tissue

LASIK EYE SURGERY



- needed to reshape the cornea
- The corneal flap is then replaced

⇒ 2.3 Loss of kidney function: dialysis

o DIALYSIS

- The preferred treatment in most cases is a kidney transplant
- However, transplant organs need to be carefully matched to recipients to prevent host immune responses destroying the introduced organ
- People with kidney failure often have to wait a number of years before a transplant is available
- Some people never get this option
- Kidney dialysis is a life-prolonging treatment
- Most common form is haemodialysis
- It must be done at least 3 times per week, with each session lasting for 4-5 hours
- It involves the patient's blood being pumped through a dialyser
- Within the dialyser, the blood passes through many tiny tubes made of semi-permeable membranes. These tubes are constantly surrounded by a special fluid called dialysate
- The semi-permeable membrane allows small substance (salt, water and urea) to pass via diffusion between blood and dialysate
- It does not allow large substances (red and white blood cells) to pass
- Dialysers allow blood to be cleaned and helps regulate the homeostasis of water and ion concentration

3. THE EFFECTIVENESS OF A TECHNOLOGY THAT IS USED TO MANAGE AND ASSIST WITH THE EFFECTS OF A DISORDER

⇒ *3.1 Hearing loss*

- o Involves both getting the right technology for the type and working with medical specialists to help each person's brain make sense of the new information

BENEFITS	LIMITATIONS
<p><u>COCHLEAR IMPLANTS</u></p> <ul style="list-style-type: none">- Hear the conversation and environmental sounds at comfortable listening levels- Detect and identify sounds in the environment, such as doorbell, car horns and the telephone- Keep their vocal loudness at an appropriate level- Understand others more accurately and with less effort- Understand speech by listening alone- Use the telephone- Enjoy and appreciate music	<p><u>COCHLEAR IMPLANTS</u></p> <ul style="list-style-type: none">- Problems can include bleeding, infections and side effects after procedure- Nerve injuries may damage taste- Nerve damage can cause weakness or paralysis in the face- Dizziness or balance problems- Loss of hearing you have left- Ringing in the ears- Leaks of the fluid around the brain- The device may not work or can get infected which means it must be removed and replaced
<p><u>BONE CONDUCTION</u></p> <ul style="list-style-type: none">- Amplifies natural movement of ossicles, making it easier to hear- Allows for better hearing in older ages- The implant only requires inner ear and cochlear nerve to be functioning	<p><u>BONE CONDUCTION</u></p> <ul style="list-style-type: none">- Expensive- Sound quality is not 100%- Surgical intervention can be very invasive
<p><u>HEARING AIDS</u></p> <ul style="list-style-type: none">- Increase the volume of sound waves entering the ear canal, thus improving various hearing loss conditions- Can be adjusted to selectively detect frequencies for which hearing loss has occurred	<p><u>HEARING AIDS</u></p> <ul style="list-style-type: none">- Don't block out background noise- They don't separate speech and noise in noisy environments- Relatively expensive- Don't allow people to hear sounds at a distance- Cannot help patients who are profoundly deaf

⇒ 3.2 Visual disorders

BENEFITS	LIMITATIONS
<p><u>SPECTACLES</u></p> <ul style="list-style-type: none"> - Provide accurate and predictable vision correction - Reliably correct your vision - Glasses are less expensive than contact lenses or surgery - They are easier to take care of than contact lenses - Glasses are unlikely to cause side effects because they don't actually touch the eye - Glasses are available everywhere and can be changed easily as vision changes - Glasses can protect eyes from injury <p><u>LASER SURGERY</u></p> <ul style="list-style-type: none"> - Procedure completed within 30 minutes or less - Very low amount of pain - No bandages or stitches and a short recovery period - Cost efficient in the long-term - More than 8/10 people don't need glasses or contact lenses for most activities after surgery 	<p><u>SPECTACLES</u></p> <ul style="list-style-type: none"> - In some types of work or in active sports, eyeglasses may be unacceptable - Some people find glasses inconvenient, uncomfortable and annoying - Eyeglasses can be broken or lost <p><u>LASER SURGERY</u></p> <ul style="list-style-type: none"> - Cost of laser - Surgery has negative side effects e.g. temporary visual disturbance - Patients need normally shaped corneas of good thickness - Visual disorder may not be completely corrected - People with autoimmune disorders or diabetes can't do the procedure - Chances of better vision based on how good vision was before surgery - Takes 2-3 months to regain full sight

⇒ 3.3 Kidney function

BENEFITS	LIMITATIONS
<p><u>DIALYSIS</u></p> <ul style="list-style-type: none"> - Helps with kidney function for those who had bad functioning kidneys - Peritoneal dialysis allows for easy transport or equipment unlike haemodialysis - Longer life expectancy 	<p><u>DIALYSIS</u></p> <ul style="list-style-type: none"> - Long process (3-5 hours three times a week) - Inhibits active lifestyles - Expensive - Diet and amount of fluid you drink needs to be restricted

