

BIOLOGY

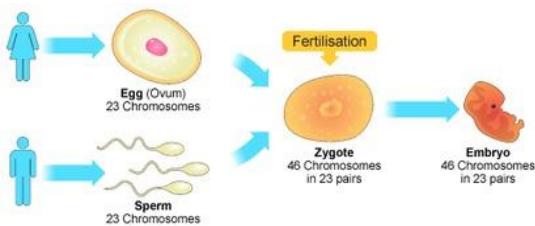
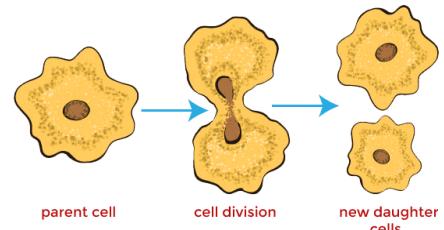
MODULE 5:

HEREDITY

5.1 Reproduction

Inquiry question: How does reproduction ensure the continuity of a species?

Asexual reproduction = a type of reproduction by which offspring arise from a single organism, and inherit the genes of that parent only. It does not involve the fusion of gametes.

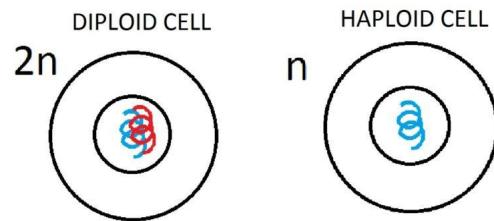


Sexual reproduction = the production of new living organisms by combining genetic information from two individuals of different types (sexes). It involves the fusion of a male gamete with a female gamete.

Gamete (sex cells) = the male or female reproductive cell that contains half the genetic material of the organism. E.g. human sperm and ova

Haploid cells contain half the number of chromosomes.
Only one copy of each chromosome.

Diploid cells contain the full set of chromosomes.



Advantages and disadvantages of asexual and sexual reproduction

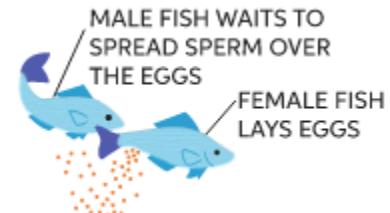
	Advantages	Disadvantages
Asexual	<ul style="list-style-type: none">- Faster- No need for another organism to be involved- When conditions are favourable can spread faster	<ul style="list-style-type: none">- No genetic variation- Bad genetic mutations build up in the population
Sexual	<ul style="list-style-type: none">- Creates genetic variation making it more likely for the species to survive if conditions change.	<ul style="list-style-type: none">- Slower- Need to find a mate (animals)- Flowers rely on pollination

Fertilisation - external and internal

Fertilisation = the fusion of male and female gametes to form a zygote.

External fertilisation = is the fusion of gametes that parents have discharged into the environment. Most fish and amphibians use external fertilisation. The eggs must be ripe for fertilisation when the sperm comes in contact with them.

Internal fertilisation = is when sperm are deposited in or close to the female reproductive tract and gametes unite within the female's body. Most of the time, internal fertilisation needs copulation (or sexual intercourse) to happen.



Zygote = fertilised egg

Why is it important for offspring to have the right number of chromosomes?

A change in the number of chromosomes can lead to problems with growth, development, and function of body systems.

Pregnancy in mammals

Mammals:

- endothermic
- have fur or hair
- suckle their young

Types of mammals:

1. Placental mammals
 - The embryo grows inside the uterus.
 - Pregnancy is longer so the embryo is born relatively well developed.
 - The placenta * in the uterus provides nutrients and oxygen and wastes are removed
 - E.g. humans, cats and whales.
2. Marsupials
 - Have a placenta; however give birth to very small, undeveloped young, which continue to develop often in a pouch.
 - E.g. kangaroo, wallaby.
3. Monotremes
 - Lay eggs from which a puggle (tiny baby) emerge
 - Develop further outside the egg being suckled by the mother

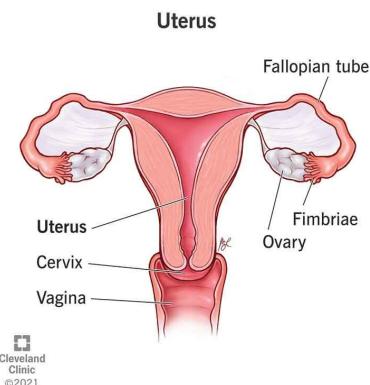
- Platypus and echidna.

* The placenta is an organ that develops in the uterus during pregnancy. This structure provides oxygen and nutrients to the growing baby and removes waste products from the baby's blood. The placenta attaches to the wall of the uterus, and the baby's umbilical cord arises from it.

Ovulation and hormone control

The menstrual cycle is the period during which an ovum (egg) matures, moves along the fallopian tubes, and enters the uterus.

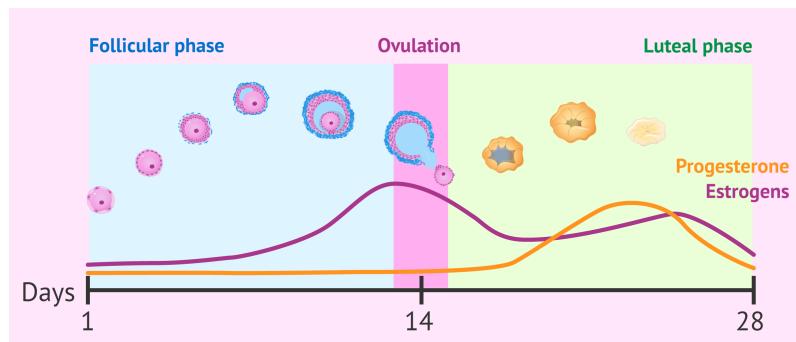
Hormones from the ovaries prepare the endometrium (uterine lining), making it thicker. If the egg is fertilised, it can embed itself in the lining. If not, the lining is shed. This shedding marks day one of the menstrual cycle, which lasts about 28 days.



Follicular phase = development of an ovum-containing follicle.

Ovulation = the rupture of the follicle and the release of the ovum.

Luteal phase = the formation and disintegration of the corpus luteum, a hormone-releasing structure.



Hormones involved in ovulation and pregnancy

GnRH - gonadotropin releasing hormone – triggers the pituitary gland to release FSH and LH

FSH – follicle-stimulating hormone

LH – luteinizing hormone - causes the egg to burst out of the follicle

Oestrogen- stimulates thickening of the endometrium.

Progesterone - maintains thickness of the endometrium

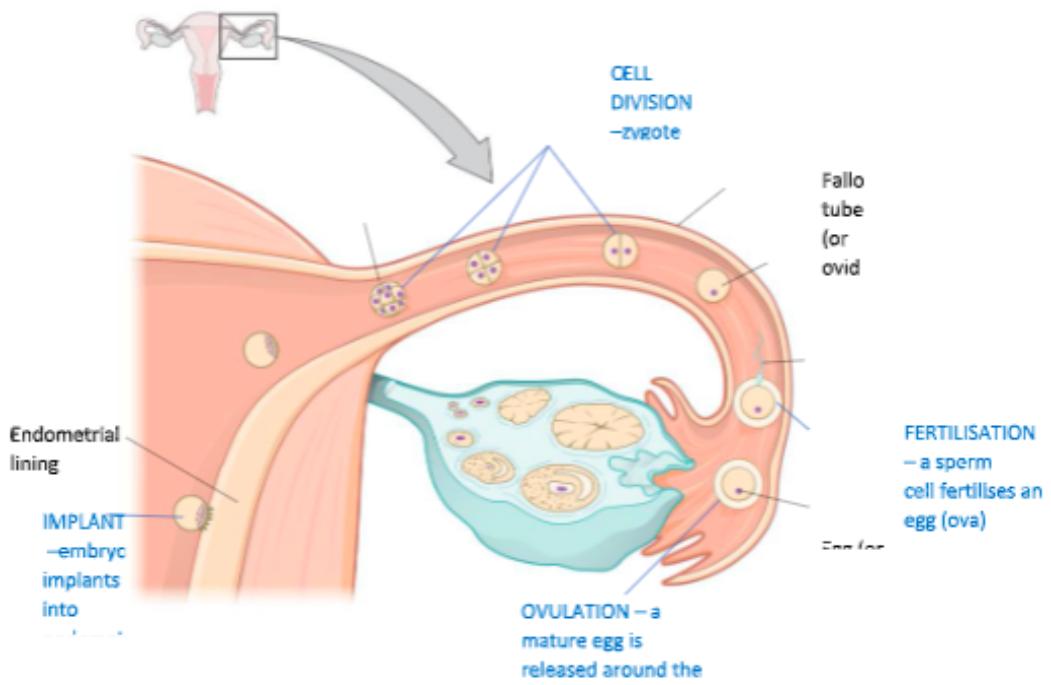
HCG – Human chorionic gonadotropin- released by the embryo to sustain the corpus luteum.

Hormone	Made by	Target	Function
FSH	Pituitary	Ovaries	Promotes development of follicle and secretion of oestrogen.
LH	Pituitary	Ovaries	Promotes ovulation, development of corpus luteum, and secretion of progesterone
Oestrogen	Ovaries	Body	Promotes menstrual cycle, development of female features and behaviour.
Progesterone	Ovaries	Uterus	Prepares uterus for, and maintains, pregnancy.
GnRH	Hypothalamus	Pituitary	Causes pituitary gland to release FSH and LH.
HCG	Embryo	Ovaries	Maintains corpus luteum for production of progesterone and oestrogen, stopping ovulation and maintains uterus lining.

Fertilisation and Implantation

Implantation:

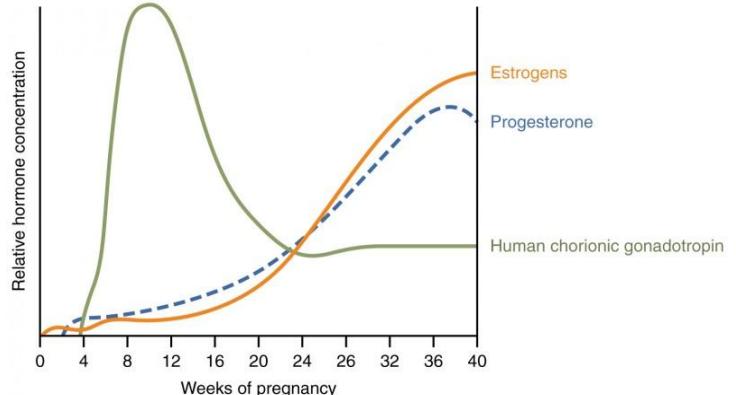
1. After fertilisation, the zygote grows by mitosis and travels down the fallopian tube to the uterus.
2. When it reaches the uterus and attaches to the lining the clump of cells is called a **blastocyst**
3. The outer cells, the **trophoblast** cells, start the formation of the placenta.
4. After implantation, the embryo begins to release human chorionic gonadotropin (HCG) which sustains the corpus luteum so progesterone and oestrogen continue to be produced.



Pregnancy and birth: in the hands of hormones

First trimester

- HCG (formed by the embryo) rises rapidly
- HCG maintains the corpus luteum, allowing it to continue secreting progesterone and oestrogen.
- Progesterone and oestrogen interact with the hypothalamus and pituitary gland, causing a decrease in GnRH, FSH and LH. This prevents menstruation or ovulation occurring.
- Progesterone also stimulates changes in the mother's body. E.g. breast growth, enlargement of the uterus.



Second trimester

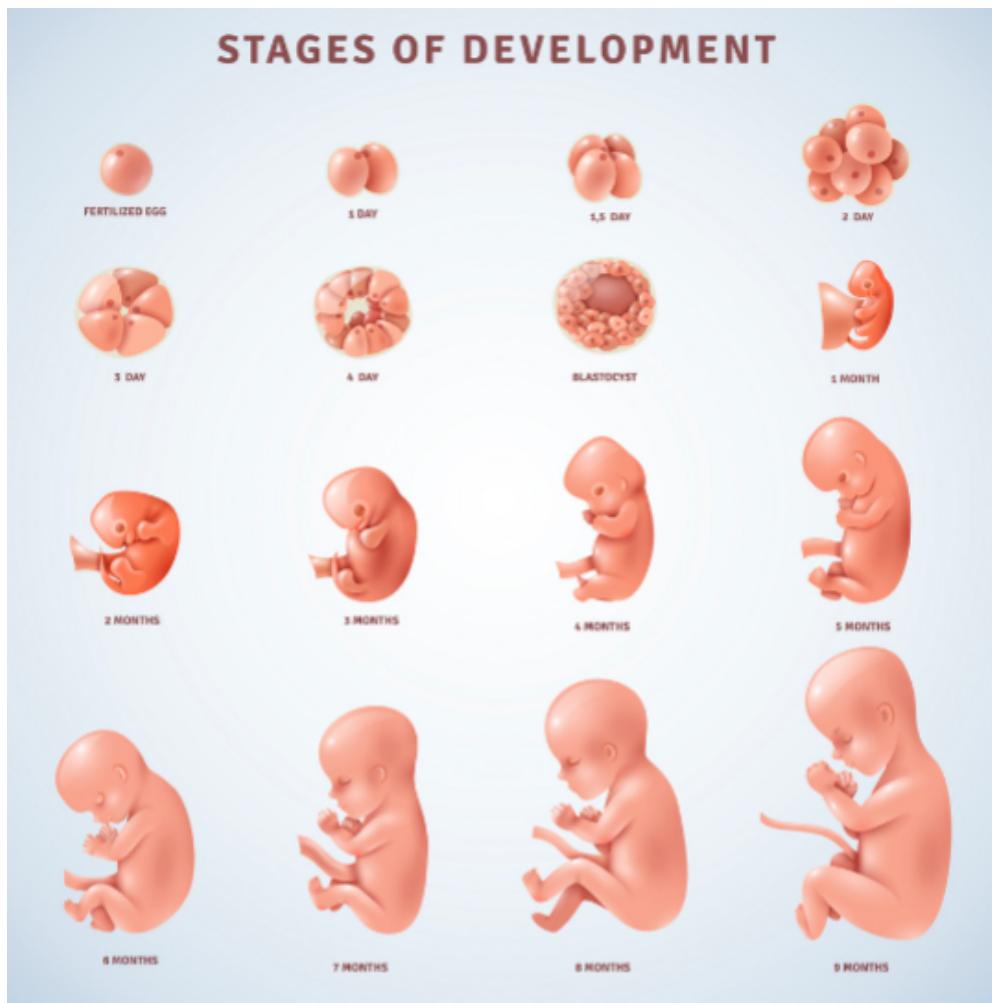
- Production of HCG declines.
- Placenta producing oestrogen and progesterone now.

Third trimester

- Oestrogen increases inducing receptors to form on the uterus wall that can bind with oxytocin.

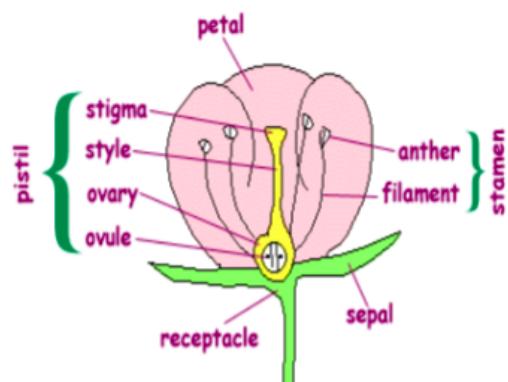
- Oxytocin is vital to trigger and maintain labour.
- Baby and mother's pituitary glands produce oxytocin during labour causing muscular contractions.

Embryo Development

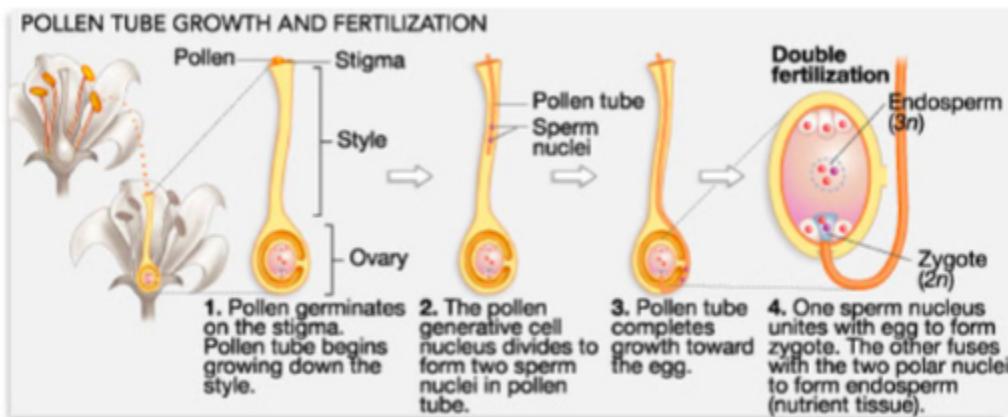


Angiosperm (flowering plant) reproduction

1. Pollen (contains male gamete) travels from the anther to a stigma.
2. Pollen germinates and a pollen tube grows down the style to the ovary.
3. The pollen generative cell nucleus divides to form two sperm cells.
4. Both sperm cells enter one of the ovules.



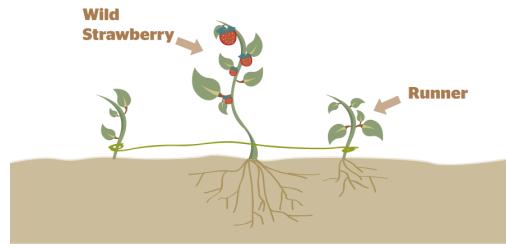
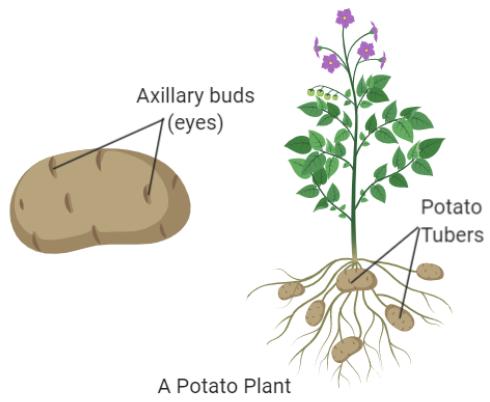
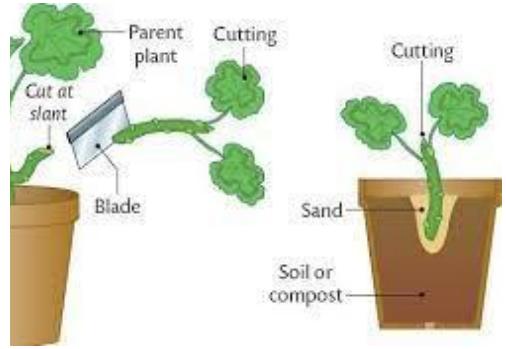
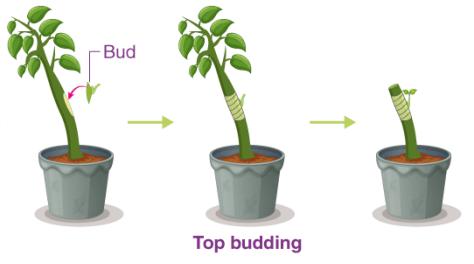
5. One fertilises the egg, the other combines with the polar nuclei to form the endosperm that provides nourishment for the zygote.
6. The ovule then matures into a seed.
7. Ovary swells into a fruit.

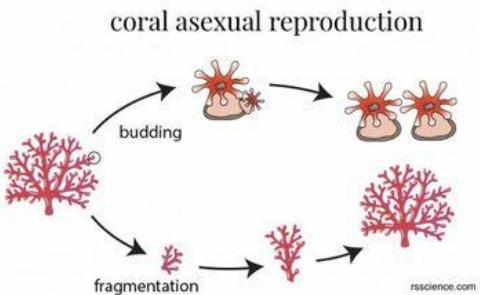
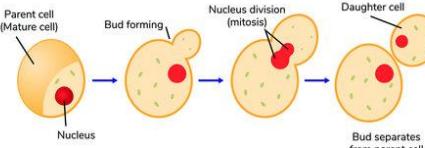
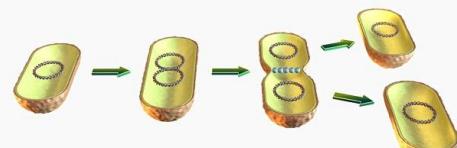
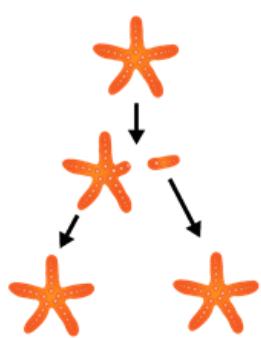


Asexual reproduction (bacteria, fungi and plants)

- Does not involve the fusion of gametes or change in the number of chromosomes.
- Offspring are genetically identical to the parent – inherit the full set of genes.
- No fertilisation. No sperm or egg. No pollination. No seed dispersal. No male and female bits involved.
- Faster than sexual reproduction when the conditions are favourable.
- Does not create genetic variation.

Method:	Process:	Example/image:
Rhizome	a stem that grows underground. It usually grows horizontally, just below the soil's surface. Since it's a stem, it has nodes and is able to put out other stems, usually straight up and above ground. This means a patch of what looks like several individual plants grouped near each other may actually all be shoots of the same plant, put up by the same rhizome.	

Runners	An above ground stem that develops horizontally along with the soil. It produces roots and aerial branches that go upward at certain points called nodes.	
Tubers	A short fleshy usually underground stem bearing minute scale leaves, each of which bears a bud in and is potentially able to produce a new plant	
Cuttings	A plant cutting is a piece of a plant that is used for vegetative (asexual) propagation. A piece of the stem or root of the source plant is placed in a suitable medium such as moist soil. If the conditions are suitable, the plant piece will begin to grow as a new plant independent of the parent, a process known as striking. A stem cutting produces new roots, and a root cutting produces new stems. Some plants can be grown from leaf pieces, called leaf cuttings, which produce both stems and roots.	
Budding in plants	Budding is inserting a single bud from a desirable plant into an opening in the bark of a compatible plant to create an advantageous variety. Budding can be used on many kinds of plants: apples, pears, peaches, and a large number of ornamentals.	

Budding in protists, coral etc	Budding occurs when a new organism grows from the body of its parents. Budding is the most common type of multiple fission in protists. The daughter nucleus is created and splits from the parent, taking some of the cytoplasm of the protist cell with it.	 coral asexual reproduction budding fragmentation <small>rsscience.com</small>
Budding in yeast	Yeast cells divide as rapidly as once every 90 min under optimal conditions. A new organism is developed from a small part of the parent's body. A bud which is formed detaches to develop into a new organism.	 Parent cell (Mature cell) Nucleus Bud forming Nucleus division (mitosis) Daughter cell Bud separates from parent cell
Binary fission in bacteria	Binary fission is a kind of asexual reproduction. It is the most common form of reproduction in lower plants such as bacteria. In this method, the nucleus splits or divides into two and then the cell splits across the middle, forming two small identical cells called the daughter cells.	
Fragmentation	A fragment of the parent breaks off and develops into an entirely new but genetically identical individual. The parent will then regenerate, or regrow, the piece that broke off, so in the end there are two new individuals from one.	

Note: vegetative reproduction, any form of asexual reproduction occurring in plants in which a new plant grows from a fragment of the parent plant or grows from a specialised reproductive structure (such as a stolon, rhizome, tuber, corm, or bulb). In many plants, vegetative reproduction is a completely natural process; in others it is an artificial one.

Reproduction in non-flowering plants

Gymnosperms



Gymnosperms
first plants to have seeds



- Any of a group of vascular plants that produce naked seeds not enclosed in an ovary.
- Use cones to reproduce.
- Have male and female cones.
- Pollen from a male cone is blown by wind onto a female cone.
- A pollen tube forms and allows the sperm cells to reach the ovule where they fertilise the ovule to form a seed.

Ferns and mosses



Fern with spores



Moss

- These plants have a life cycle involving an alternation of generations.
- They have a haploid (n) structure and a diploid ($2n$) structure.
- The gametophyte is the haploid structure.
- The sporophyte is diploid.

Reproduction in fungi

Fungi

- Yeasts, moulds and mushrooms
- Eukaryotic, heterotrophic organisms with cell walls
- Obtain food by secreting digestive enzymes and absorbing dissolved molecules.
- Do not photosynthesise
- Main decomposers in most ecosystems.

Asexual reproduction

Budding – yeasts are unicellular fungi. A bud is formed on the side of a cell, the nucleus divides and two separate, identical cells are formed.

Fragmentation – hyphae break into segments that each grow into a new thallus.

Spore formation – a specialised reproductive sac called a sporangium can result in the release of large numbers of tiny spores resulting in the widespread dispersal of fungi.

Fairy Rings



A fungal mycelium has many branching structures that grow outward in a circle as it expands underground as they seek nutrients. As this occurs, the fruiting bodies form in a circular shape above ground on the outer parts. The grass above the leading edge of the fungal mycelium becomes greener due to the fungus enriching the soil below with nutrients.

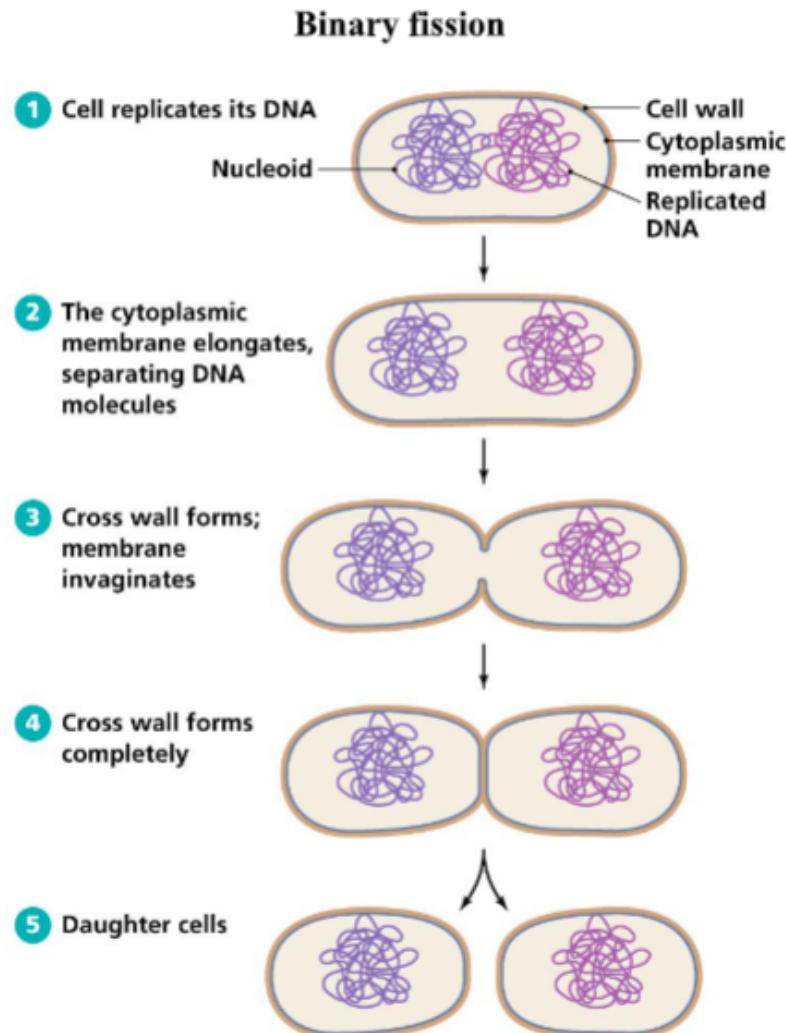
The spores allow Penicillium fungi to reproduce and so assist in the survival of their species.

[Note: Spores also assist in their dispersal and so allow them to expand their distribution and colonise new environments, which assists in species survival.]

Binary fission in prokaryotes

- Prokaryotic organisms
- Unicellular.
- No membrane bound organelles e.g. no nucleus.

- Bacteria and archaea.
- Reproduced by binary fission.



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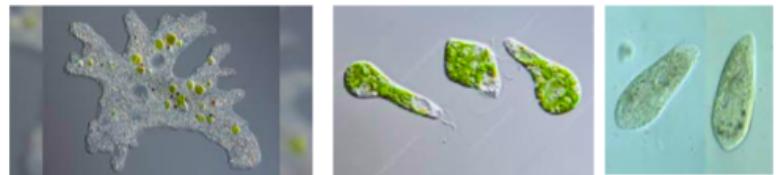
Reproduction of protists

What are protists?

All protists are eukaryotes, i.e. organisms with a nucleus. However, they are neither fungi nor plants nor animals. They are a separate kingdom of living things. Most of them are unicellular; however, a few protists are multicellular.

Asexual mode of reproduction can occur via several methods, as described below.

Binary Fission: the parent body divides into two equal daughter cells by undergoing mitosis. Examples: Amoeba, Euglena, and Paramecium.



Multiple Fission: here, the parent cell divides into several daughter cells. Examples: Amoeba and Plasmodium.

Spore Formation: some protists form spores by asexual reproduction to withstand unfavourable or undesirable environmental conditions. Once spores are exposed to the optimum conditions, they germinate and form new progeny. Example: slime moulds.

Budding: a small outgrowth or protrusion develops on the body of the parent cell which eventually pinches off to form a new organism. Example: Arcella (a sarcodine)

Protozoa are the animal-like species in the kingdom of Protista. They include many parasitic protozoa which can cause disease in humans and other organisms.

E.g. Giardia

Giardia is a tiny parasite (germ) that causes the diarrhoeal disease giardiasis. Giardia is found on surfaces or in soil, food, or water that has been contaminated with faeces (poop) from infected people or animals. (Don't swim in Manly Dam after a lot of rain)



E.g. Plasmodium

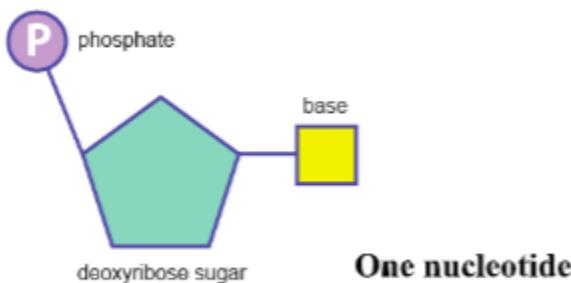
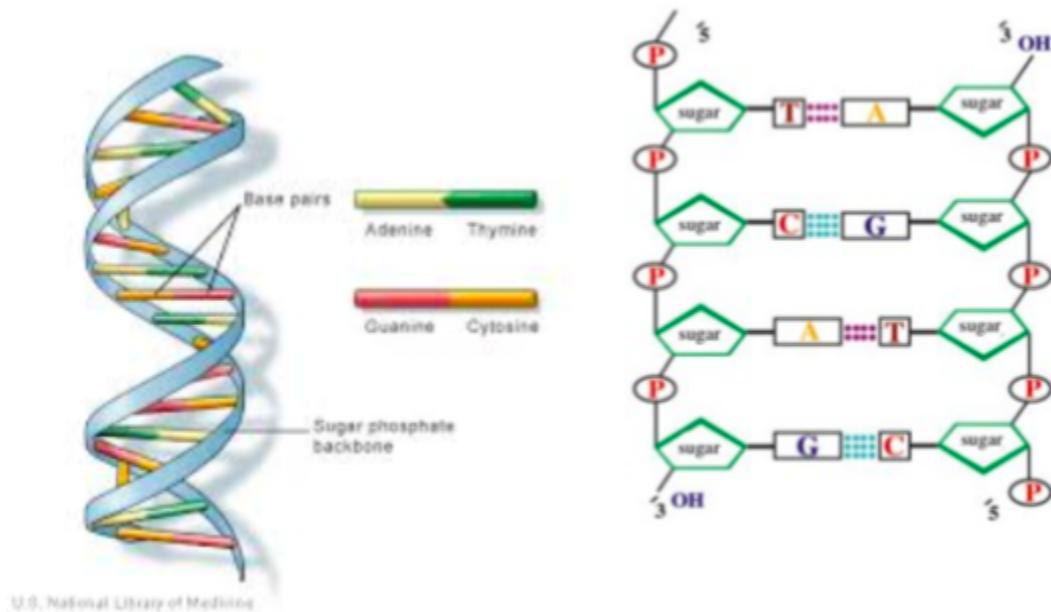
Plasmodium is the parasite that causes Malaria. Malaria is one of the most problematic diseases in many tropical countries in the world. The vector (organism that carries plasmodium) is the female Anopheles mosquito



5.2 Cell Replication

Inquiry question: How important is it for genetic material to be replicated exactly?

- DNA is made up of molecules called nucleotides.
- Each nucleotide contains a phosphate group, a sugar group and a nitrogen base.
- The bases are joined to the sugar molecule on the backbone.
- The four types of nitrogen bases are adenine (A), thymine (T), guanine (G) and cytosine (C).
- The order of these bases is what determines DNA's instructions, or genetic code.

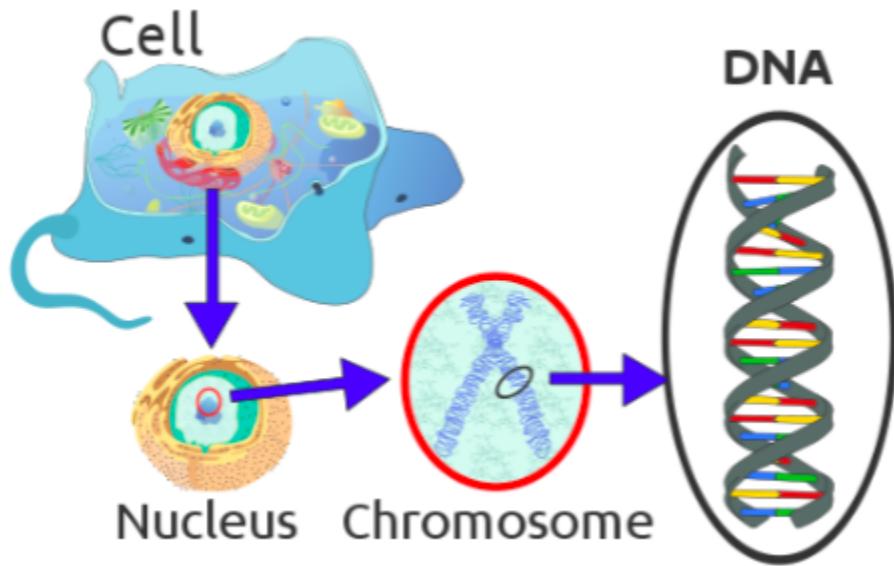


A chromosome is a long coiled DNA molecule

A gene is a section of a chromosome with specific bases along the DNA

Prokaryotic cells have their DNA as a single looped circular chromosome. Some have an additional small circle of DNA called a plasmid.

Eukaryotic cells have multiple linear chromosomes that are not looped contained in the nucleus



Discovery of DNA structure:

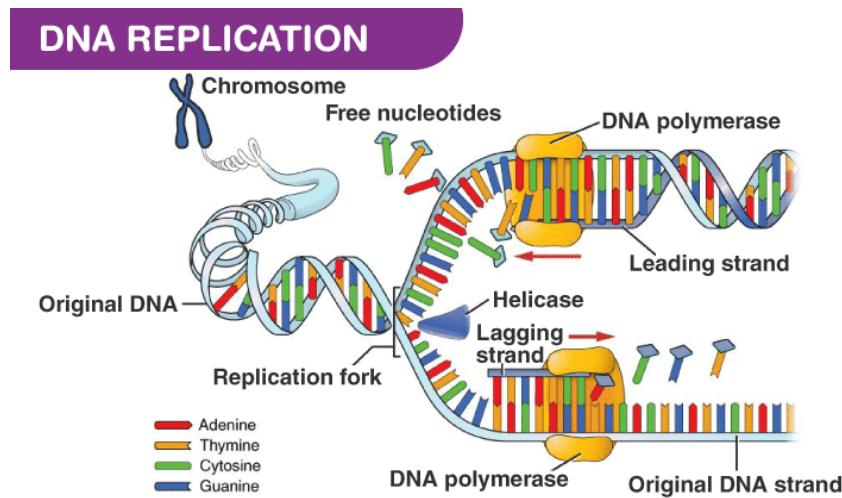
At King's College in London, Rosalind Franklin and Maurice Wilkins were studying DNA. Wilkins and Franklin used X-ray diffraction as their main tool -- beaming X-rays through the molecule yielded a shadow picture of the molecule's structure, by how the X-rays bounced off its component parts.

DNA Replication

DNA replication is essential for cell division to provide identical genetic material in each new daughter cell.

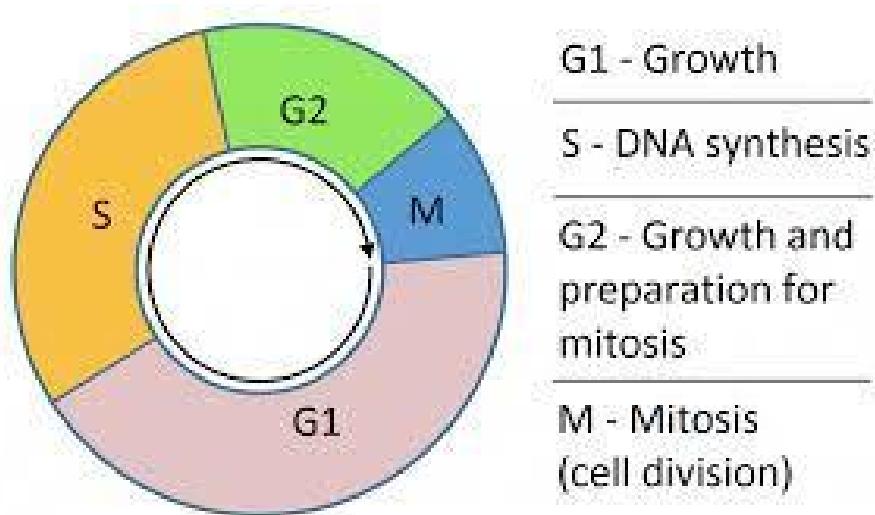
1. With the help of the enzyme DNA helicase, the double helix unwinds to form two strands.
2. New nucleotides are transported towards these strands and link to them according to the base-pairing rule.
3. DNA polymerase is the enzyme responsible for building the new strand.
4. When fully replicated the two new double strands wind back up into the double-helix form.

5. DNA ligase is the enzyme joining it all together.
6. When the two new cells are formed in mitosis, each has an identical copy of the DNA structure in its nucleus.



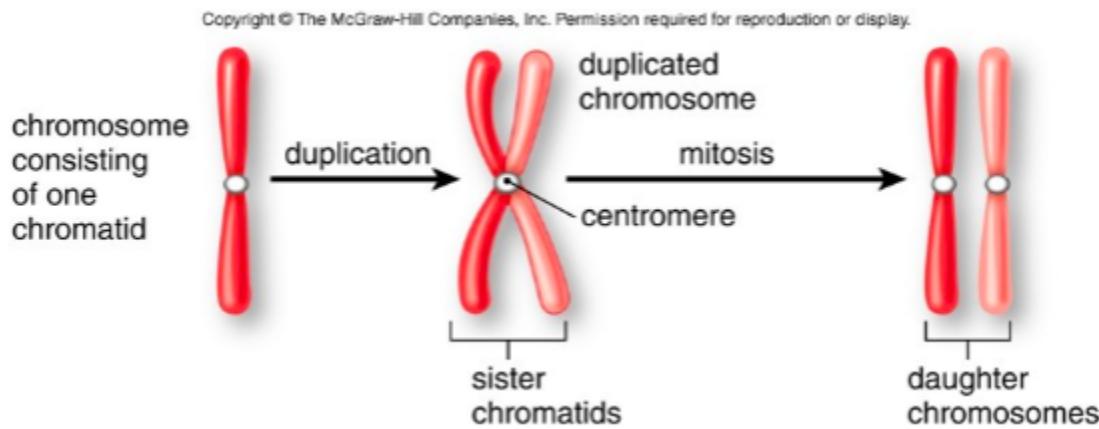
The Cell Cycle

- Cells grow and replicate in four main phases.
- Interphase consists of the G1, S and G2 phases. This is where the cell is growing by producing proteins and more organelles.
- They mature in the G1 phase.
- DNA replication (essential for cell division) occurs in the S phase.
- They prepare for division in the G2 phase.
- Cell division (mitosis) occurs in the M phase.



Chromosomes and chromatids

- During the G1 phase, chromosomes exist as a single chromatid, made of a single DNA molecule.
- During the S phase, DNA replication occurs. This results in each chromosome being made of two identical chromatids. Each chromatid is made of a single DNA molecule. The two DNA molecules are joined together at the centromere.
- During the M phase, cell division occurs by mitosis. The two chromatids separate into two identical daughter chromosomes.

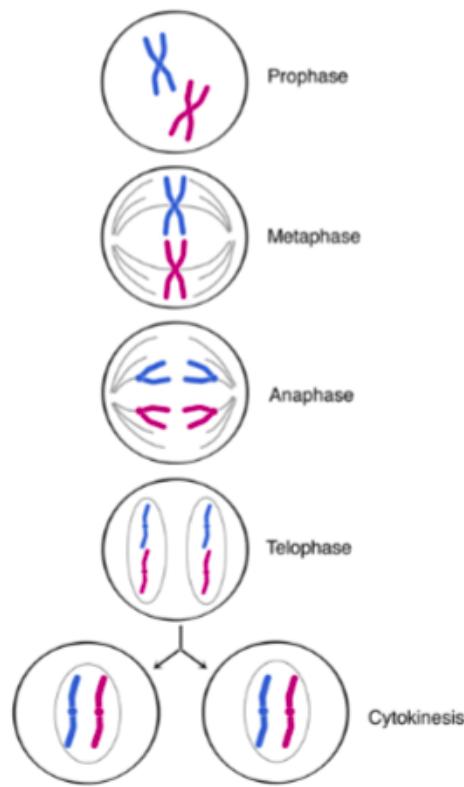
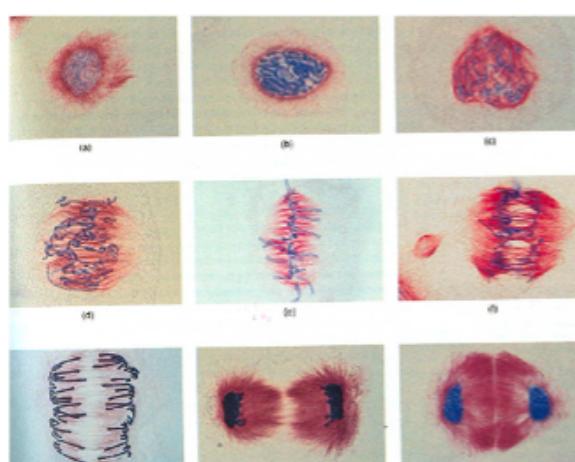


Mitosis

Mitosis (cell division) consists of 4 phases PMAT
(DNA replication has occurred prior to mitosis)

1. Prophase (pro = before)
2. Metaphase (m = middle)
3. Anaphase (a = away)
4. Telophase (t = two)

After telophase the cell undergoes cytokinesis. This is where the cytoplasm divides, resulting in two identical daughter cells.



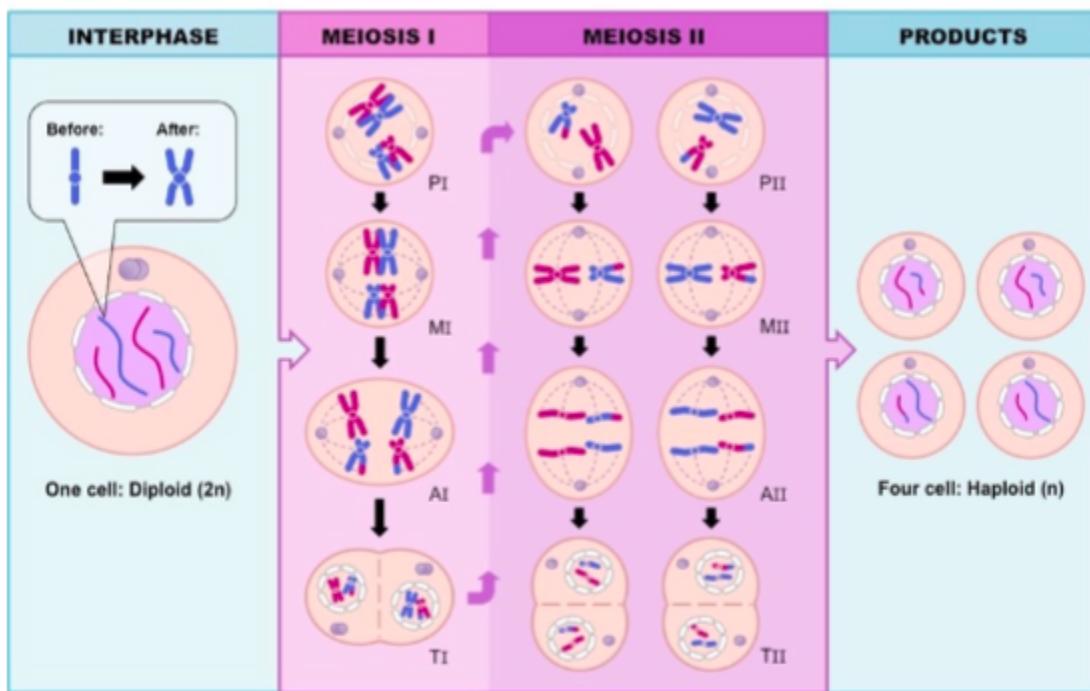
Meiosis

Key terms:

Germ cells = gametes (sex cells) → haploid

Somatic cells = body cells → diploid

- Cell division creating gametes (sex cells)
- One diploid ($2n$) cell undergoes two stages of meiosis resulting in four haploid (n) cells.
- All body cells contain two copies of each chromosome.
- Gametes contain one of each chromosome.
- Humans diploid number = 46
- Human haploid number = 23
- Sperm (23) fertilised ova (23) = diploid cell of 46 chromosomes.



(Meiosis 2 is just like mitosis)

centrioles

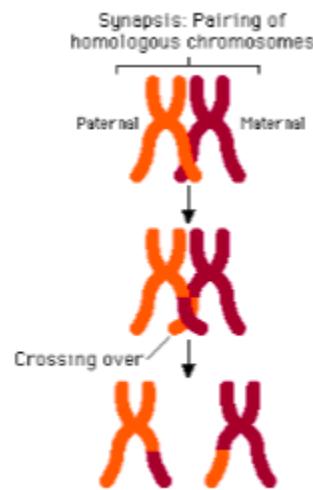
Causes of variation in meiosis

1. Independent assortment = The way the chromosomes line up at metaphase is not dependent on other pairs.
Random segregation = Random chromatids separate into the newly forming egg/sperm.
 - Chromosome pairs line up and separate independently of each other.

- Homologous pairs are divided between separate gametes during meiosis
- 46 chromosomes randomly segregate to produce a possible 2^{23} different gametes.

2. Crossing over

- Adjacent chromatids twist around each other, split, and re-join to form new combinations of genetic material.



Mitosis vs Meiosis

	Mitosis	Meiosis
<i>Reproduction</i>	Asexual	Sexual
<i>Products</i>	2 diploid cells	4 haploid cells
<i>Cell type</i>	germ cells (gametes/sex cells)	somatic (body cells)

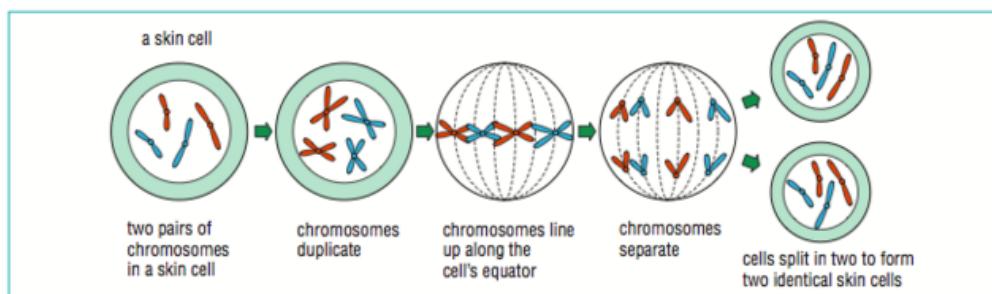
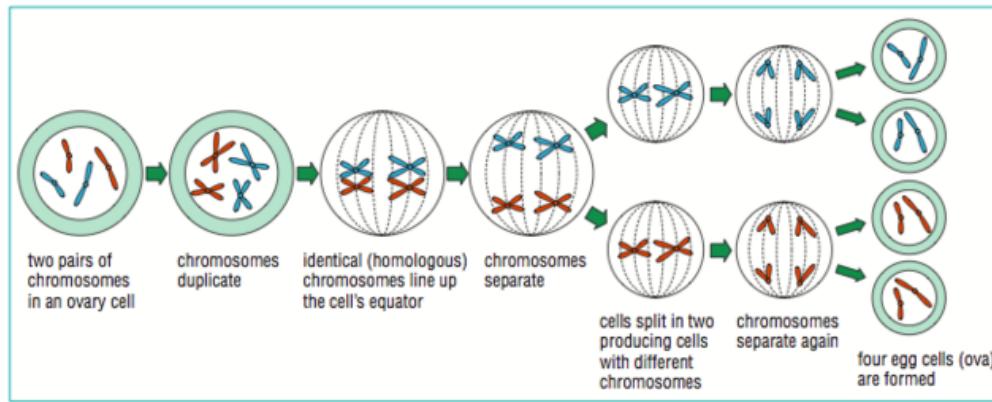


Fig 3.1.6 Mitosis produces cells that are identical to the parent cell. Mitosis occurs in all cells except sperm and egg cells.



5.3 DNA and Polypeptide Synthesis

Protein Synthesis

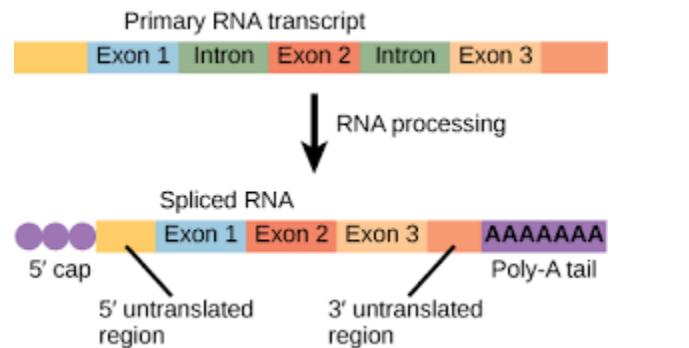
TRANSCRIPTION

1. RNA polymerase is the enzyme assisting transcription.
2. The double helix of DNA separates.
3. Free nucleotides are attracted according to the base code G-C, A-U. This forms **messenger RNA** on one side of the split DNA. (RNA has Uracil instead of Thymine)
- Editing**
4. **mRNA** moves out from the nucleus, into the cytoplasm to a **ribosome**.

TRANSLATION

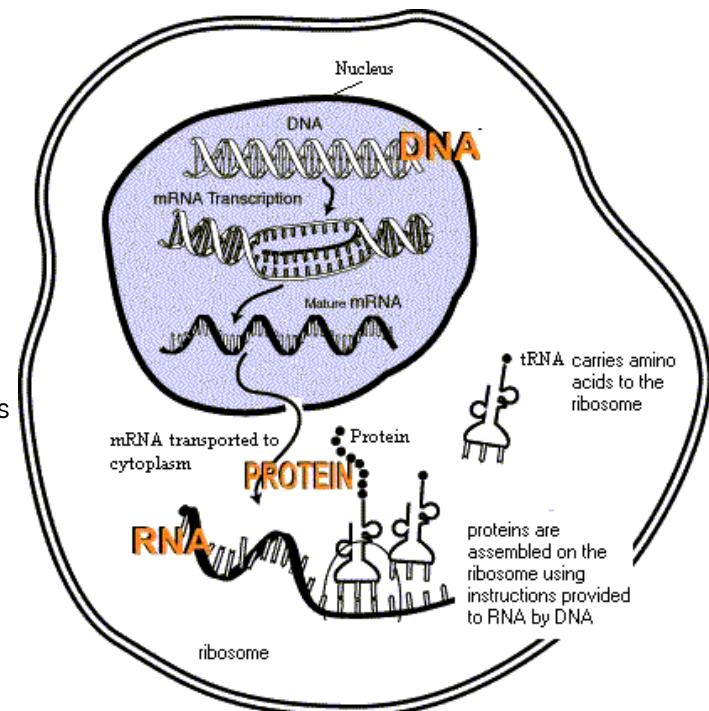
1. mRNA has arrived at the ribosome and **Transfer RNA** takes charge!
2. **tRNA** is different to mRNA as it has three bases and an **amino acid** attached.
3. The amino acid brought depends on the base code of mRNA. E.g. GGC codes for the amino acid glycine.
4. Lots of tRNA come bringing amino acids and they all join up to form a **polypeptide**. When it forms its **3D** shape, this is a **protein**.
5. Good job!!!

Editing

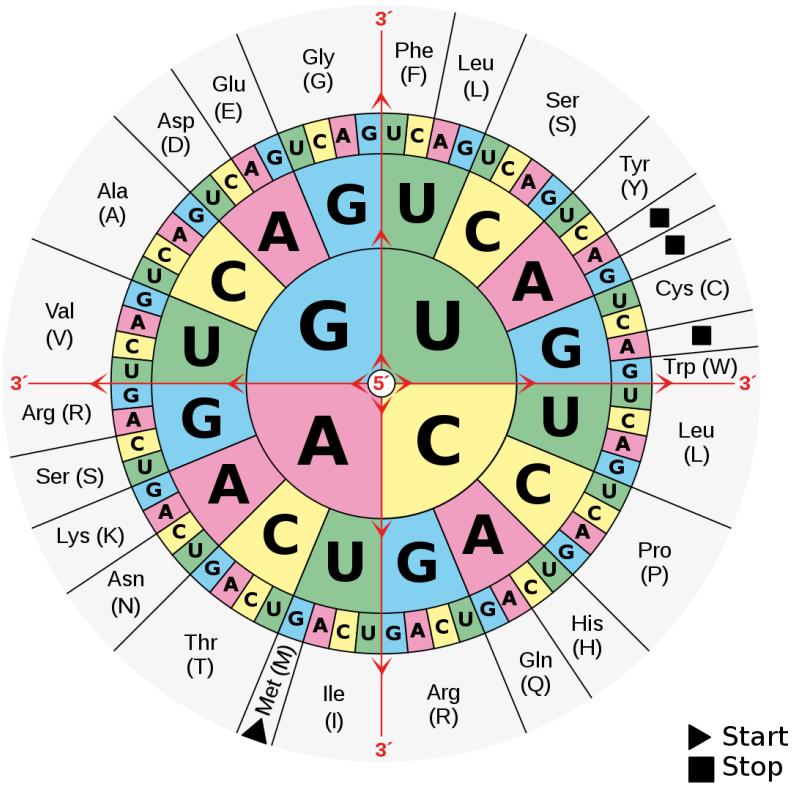


The sections of DNA (or RNA) that code for proteins are called **exons**. (**E**xtra special)

Introns are noncoding sections (not much use)



<http://www.youtube.com/watch?v=h3b9ArupXZg> (11 MINS BOZMAN)



Vocab:

Words	Meaning
<i>Transcription</i>	The process by which a cell makes an RNA copy of a piece of DNA. This RNA copy, called messenger RNA (mRNA), carries the genetic information needed to make proteins in a cell. It carries the information from the DNA in the nucleus of the cell to the cytoplasm, where proteins are made.
<i>Polypeptide</i>	A peptide is a short chain of amino acids linked by chemical bonds. A longer chain of linked amino acids is a polypeptide. The proteins manufactured inside cells are made from one or more polypeptides
<i>RNA</i>	The primary function of RNA is to create proteins via translation. RNA carries genetic information that is translated by ribosomes into various proteins necessary for cellular processes. mRNA, rRNA, and tRNA are the three main types of RNA involved in protein synthesis.
<i>mRNA</i>	Messenger RNA is a type of RNA that is necessary for protein production. In cells, mRNA uses the information in genes to

	create a blueprint for making proteins. Once cells finish making a protein, they quickly break down the mRNA.
<i>RNA polymerase</i>	a multi-unit enzyme that synthesises RNA molecules from a template of DNA through a process called transcription
<i>Gene</i>	Made up of DNA
<i>Exons</i>	a region of the genome that ends up within an mRNA molecule
<i>Introns</i>	An intron is a region that resides within a gene but does not remain in the final mature mRNA molecule following transcription of that gene and does not code for amino acids that make up the protein encoded by that gene
<i>Translation</i>	the process through which information encoded in messenger RNA (mRNA) directs the addition of amino acids during protein synthesis
<i>tRNA</i>	Transfer RNA (abbreviated tRNA) is a small RNA molecule that plays a key role in protein synthesis. Transfer RNA serves as a link (or adaptor) between the messenger RNA (mRNA) molecule and the growing chain of amino acids that make up a protein.
<i>Codon</i>	A codon is a DNA or RNA sequence of three nucleotides (a trinucleotide) that forms a unit of genomic information encoding a particular amino acid or signalling the termination of protein synthesis (stop signals). There are 64 different codons: 61 specify amino acids and 3 are used as stop signals.
<i>Anticodon</i>	a triplet of nucleotide bases in transfer RNA that identifies the amino acid carried and binds to a complementary codon in messenger RNA during protein synthesis at a ribosome.

Create a table to compare the processes of **DNA replication** and **protein synthesis**.

Similar and differences

Feature	DNA replication	Protein synthesis
First Step	Enzyme assists unzipping of DNA double helix	Enzyme assists unzipping of DNA double helix
Bases involved	Guanine - cytosine Adenine - Thymine	Guanine - Cytosine Adenine - Uracil
Location it occurs	Nucleus	Nucleus → Cytoplasm → Ribosome

End result	Two strands of DNA molecules identical to the original	Polypeptides that fold into 3D shape = protein
Purpose	Reproduction and continuity of species	Reproduction and continuity of species

Chromosomes, genes and alleles

Chromosome = a long piece of DNA wound up around a protein. Each chromosome contains many genes.

Genes = small sections of DNA that code for proteins. They contain the instructions for our individual characteristics.

Alleles = the different variations of a gene found at the same place on a chromosome

- e.g the gene for eye colour in humans has alleles brown, blue, green etc.

Genotype → born with genes e.g blonde hair

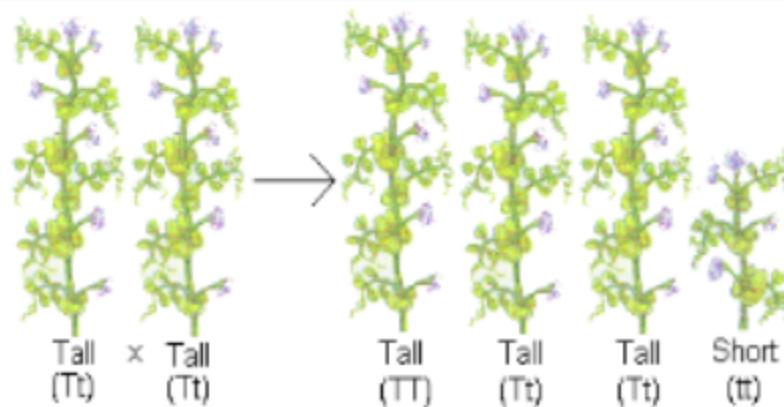
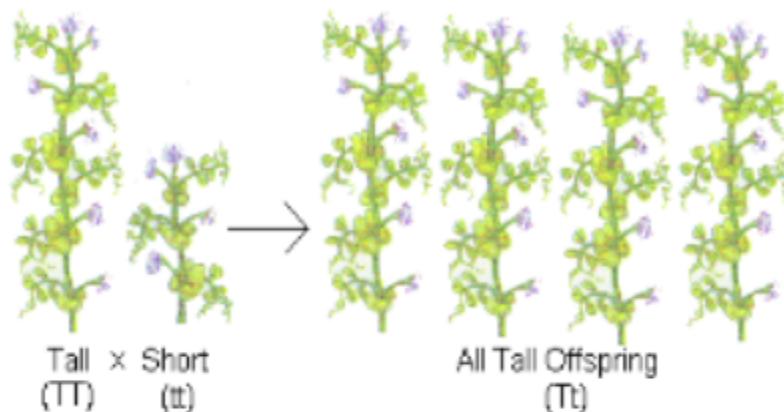
Phenotype → environment affecting e.g dying hair brown

5.4 Genetic Variation

Introduction to Genetics

Genetics is a field of Biology that looks at genes and the ways they are inherited

MENDEL'S FAMOUS 3:1 RATIO



TT – purebred tall plant (homozygous)

tt - purebred short plant (homozygous)

Tt – hybrid tall plant (heterozygous) – tall plant carrying gene for short plant

MENDEL'S CONCLUSIONS

- Each pure-breeding plant had two identical copies of a hereditary factor. eg TT & tt.
- Only one factor goes into each sperm or egg.
- Mendel identified them as 'factors' we now call them_____.
- There must be a dominant gene and a recessive gene.
- In this case tall is dominant and short is recessive.
- Alleles – forms of genes –types of genes for a characteristic. Eg. tall and short pea plants
E.g. blue or brown alleles for the gene of eye colour
- Individuals that have inherited identical genes are called homozygous or pure breeders.
- TT-homozygous tall.
- Individuals with different alleles are called heterozygous or hybrid Tt – heterozygous tall
- **Genotype – is the genetic composition eg. TT, tt, Tt.**
- **Phenotype – refers to characteristic shown by the individual eg. Tall, short**

Punnett Squares

	B	b	
Dominant → B	BB	Bb	← Heterozygous
Recessive → b	Bb	bb	← Homozygous

Incomplete and Codominant genes

INCOMPLETE DOMINANCE

- One allele is not completely dominant over another.
- The heterozygous form (RW) has a different phenotype to both homozygous forms.
e.g Snapdragon flowers red x white = pink



e.g.



CODOMINANCE

- Both alleles are expressed.
- Roan cattle Red bull x white cow = roan cow (both red and white hair)

Genes with multiple alleles

Blood Groups

A and B are codominant and O is recessive

Rhesus positive and rhesus negative blood. Rhesus positive blood has a molecule (antigen) on the surfaces of red blood cells. Rhesus negative does not.

If a R. negative person receives R positive blood, their immune system will target the red blood cells, because the red blood cells have a 'foreign' unrecognisable molecule attached.

(Your immune system (white blood cells) attack anything that is not recognisable as 'self'. E.g. bacteria, foreign proteins etc.)

Rhesus positive people can be given rhesus negative blood as there is no unrecognisable molecule attached.

O negative is the 'universal blood type' because everyone can receive this type.

Genotype	Blood group (phenotype)
AA	A
AO	A
BB	B
BO	B
AB	AB
OO	O

The ABO Blood System

Blood Type (genotype)	Type A (AA, AO)	Type B (BB, BO)	Type AB (AB)	Type O (OO)
Red Blood Cell Surface Proteins (phenotype)	A agglutinogens only	B agglutinogens only	A and B agglutinogens	No agglutinogens
Plasma Antibodies (phenotype)	b agglutinin only	a agglutinin only	NONE	a and b agglutinin

It takes *all* types.

TYPE	YOU CAN GIVE BLOOD TO	YOU CAN RECEIVE BLOOD FROM
A+	A+, AB+	A+, A-, O+, O-
O+	O+, A+, B+, AB+	O+, O-
B+	B+, AB+	B+, B-, O+, O-
AB+	AB+	EVERYONE
A-	A+, A-, AB+, AB-	A-, O-
O-	EVERYONE	O-
B-	B+, B-, AB+, AB-	B-, O-
AB-	AB+, AB-	AB-, A-, B-, O-

Sex-linked Inheritance

SEX LINKED CHARACTERISTICS

PROBLEM

Colour blindness in humans is sex linked. The gene for colour blindness or Normal vision is carried on the X chromosome.

$X^N Y$ = Normal vision male

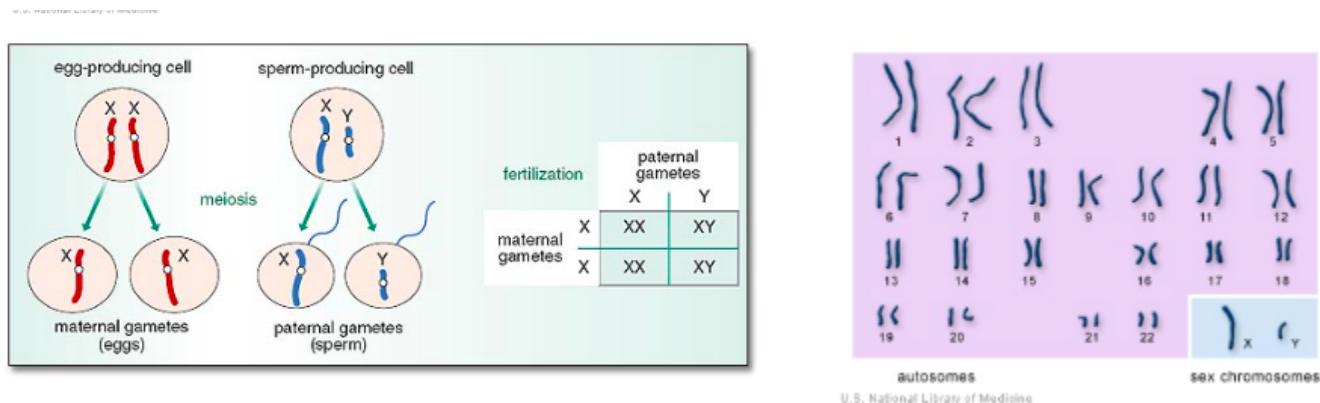
$X^n Y$ = Colour blind male

$X^N X^N$, $X^N X^n$ = Normal vision female

$X^n X^n$ = Colour blind female

If given a problem:

1. Determine the possible genotypes and phenotypes of the offspring of a normal vision male and a heterozygous normal vision female.
2. Show (with a Punnett square) how a colour blind female offspring could be produced.



Pedigrees (family trees)

How to find which type of pedigree it is:

- autosomal dominant:
 - an affected individual ALWAYS has at least one affected parent
 - Two affected parents can have an unaffected child
- autosomal recessive:
 - an affected individual may have unaffected parents
 - two affected parents only have affected children
 - not on sex chromosome
- X chromosome recessive
 - occurs more often in males
 - male inheritance can skip a generation (e.g grandfather to grandson)
 - all sons of an affected female are also affected.

Module 6:

Genetic

Change

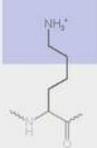
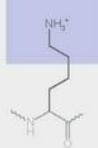
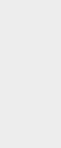
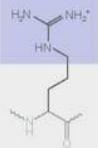
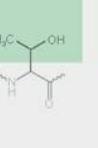
6.1 Mutations

Introduction to mutations

Types of genetic mutations:

Point mutation: involved alteration to one (or sometimes a few) of the bases, there are three types:

- silent - there is no change to the amino acid sequence in the polypeptide
- missense (substitution) - there is a change to one amino acid in the polypeptide
- nonsense - there is a premature stop codon, which shortens the polypeptide and so usually results in a non-functional protein.

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

How to discover what type of mutation it is:

1. change into mRNA (its matching pairs)
2. create amino acids using given table, until it says stop
3. identify difference in the mutated DNA
4. repeat steps 1 and 2 for the mutated DNA
5. decide what type of mutation it is depending on if it has shifted, or one amino acid has changed etc.

Causes of mutations

Mistakes can occur during DNA replication. These are common, however there are cellular DNA repair mechanisms that 'proof-read' DNA after replication and correct many of these mutations.

Effects of mutations

Somatic cell (body cells) mutations affect body cells

- Mutation might have no effect.
- Cell not able to function properly - cell death.
- Malfunction in cell – becomes cancerous
- Future generations are not affected.

Germ-line (sex cells) mutations occur in gametes.

- New inheritable genes may be detrimental to survival, an improvement to survival or neither good or bad.

Mutagens

Mutagens – external causes of mutations, including chemicals and radiation.

E.g. agent orange, UV radiation.

Radiation	Chemicals	Infectious Agents
UV (from sunlight)	X-rays (medical uses)	Carcinogens (e.g. cigarettes)
		Processed foods & preservatives
		Cosmetics & cleaning products
		Viruses (e.g. HPV)
		Bacteria (e.g. Helicobacter)

Point mutations

(same as above) + insertion or deletion of bases in a DNA strand. It is called a **frameshift**.

We work these questions out the same as above but instead a base will be deleted OR added, changing the sequence and therefore amino acids.

Original DNA TACGTTGCGAAATTCACT

Mutated DNA TACGTTAGCGAAATTCACT

Second Position				Third Position									
First Position		U		C		A		G		U		C	
U	C	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid	code	Amino Acid
U	A	UUU	phe	UCU		UAU	tyr	UGU	cys				
		UUC		UCC	ser	UAC		UGC					
		UUA	leu	UCA		UAA	STOP	UGA	STOP				
		UUG		UCG		UAG	STOP	UGG	trp				
C	G	CUU		CCU		CAU	his	CGU					
		CUC	leu	CCC		CAC		CGC					
		CUA		CCA	pro	CAA	gln	CGA					
		CUG		CCG		CAG		CGG					
A	T	AUU		ACU		AAU	asn	AGU					
		AUC	ile	ACC		AAC		AGC					
		AUA		ACA		AAA	lys	AGA					
		AUG	met	ACG		AAG		AGG					
G	C	GUU		GCU		GAU	asp	GGU					
		GUC		GCC		GAC		GGC					
		GUА	val	GCA		GAA	glu	GGA					
		GUG		GCG		GAG		GGG					

Examples of single-gene mutations

Sickle cell anaemia

People with sickle cell anaemia produce abnormal haemoglobin proteins resulting in red blood cells in the shape of a sickle. The main function of red blood cells is to carry oxygen, attached to haemoglobin, around the body. This is caused by a single gene mutation as shown in the diagram below.

$H^N H^N$ = normal red blood cell

$H^S H^S$ = sickle shaped red blood cell (Unable to carry as much oxygen)

$H^N H^S$ = both normal and sickle shaped red blood cells. *

*In some tropical countries it is an advantage to be heterozygous sickle cell, as the malaria parasite cannot live in a sickle shaped cell. In these countries, the mutant sickle allele has a high frequency in the gene pool.

Chromosomal Mutations-structural

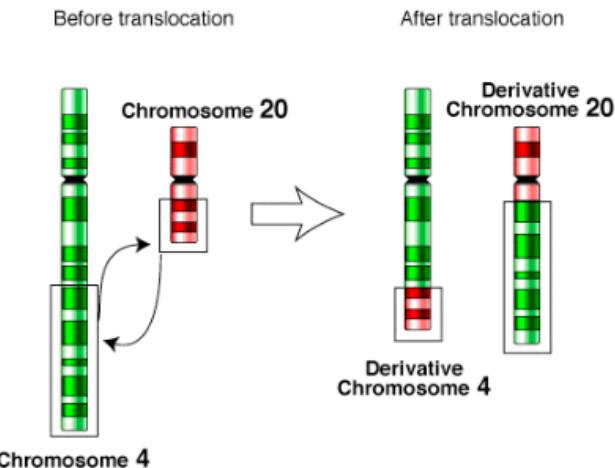
Chromosomal mutations can cause structural changes to the chromosome. This can be caused by having chunks deleted, duplicated or translocated from one chromosome to another. Chunks can also be inverted. (See diagrams below)

- Deletion
- Duplication
- Inversion
- Translocation

Two translocation mutations.

A large chunk of chromosome 4 translocated to chromosome 20.

A small chunk of chromosome 20 translocated to chromosome 4.



Balanced structural abnormalities involve no gain or loss of genes, just a rearrangement

- inversions
- translocations

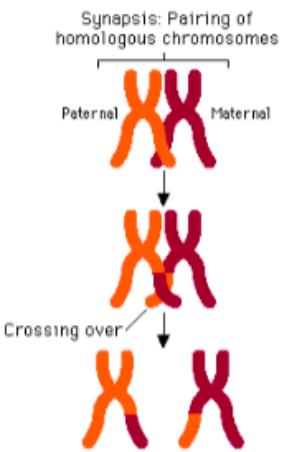
Unbalanced structural abnormalities involve gain or loss of genes

- deletions
- duplications

Crossing-over causes mutations

Crossing-over can occur between homologous chromosomes during meiosis. This is an exchange of corresponding genes between the chromosomes.

- If crossing-over does not occur in the typical manner, **recombination errors** can occur.
- A different amount of genetic material could be exchanged, resulting in one homologous chromosome with missing genetic material (deletion mutation) and the other homologous chromosome with duplication of a region.
- Crossing-over can occur between non-homologous chromosomes resulting in translocation mutations.



Chromosomal mutations-numerical 8

Missing chromosomes or **extra** chromosomes.

Nondisjunction = meiosis resulting in incorrect numbers of chromosomes in gametes.

Aneuploidy = having an abnormal number of chromosomes in a cell.

E.g. Down syndrome

A normal human karyotype (a picture of a person's chromosomes) has 46 chromosomes. 23 pairs.

Down syndrome, sometimes referred to as 'trisomy-21' is caused by a person having three copies of chromosome 21

Sex chromosome abnormalities

Triple X syndrome

Triple X syndrome, also called trisomy X or is characterised by the presence of an additional X chromosome in each of a female's cells. Although females with this condition may be taller than average, this chromosomal change typically causes no unusual physical features

Jacob's syndrome

XYY syndrome is a genetic condition in which a male has an extra Y chromosome. Symptoms are usually few. They may include being taller than average, acne, and an increased risk of learning problems.

Causes of numerical abnormalities

- Errors occurring during meiosis.
- Failure of homologous chromosomes or sister chromatids to separate correctly (nondisjunction).

Long response mutations and variation

Justify the following statement:

Germ-line mutations can be harmful, beneficial or neutral. However, if they are lethal, they increase variation and can create new alleles in the population.

ALARM:

Define:

Describe

Explain

Examples (link to question)

1. Sickle cell anaemia (bad)
2. Cystic fibrosis (bad)
3. Almond trees (good)

Justify

The term mutation is used to refer to any changes that occur in the sequence of bases within an organism. Germ-line mutation is the mutation that occurs in gametes (an egg cell or sperm), and this means it can be inherited into the offspring. These types of mutations can be beneficial as it can cause gene mutations that strengthen the environment for example, almond trees. But these types of mutations can also be harmful and cause birth deficiencies, for example sickle cell anaemia and cystic fibrosis. Both diseases/deficiencies that cannot be cured.

The almond tree mutation refers to the creation of edible almonds, when the seeds were first discovered in the wild they contained a chemical called amygdalin, which is a lethal chemical when consumed by humans. A single gene mutation in one of the wild almond seeds resulted in a new variety of almonds that no longer contained this chemical. This reflects how germ-line mutations are beneficial to society, they have successfully created a new food to eat that is extremely popular and brings in millions of dollars for Australia each year.

Sickle cell anaemia is a disease in red blood cells that contort into a sickle shape. The cells die early, leaving a shortage of healthy red blood cells, it blocks blood flow to organs depriving them of oxygen. In sickle cell anaemia, blood is also chronically low in oxygen. This lack of oxygen-rich blood can damage nerves and organs, including kidneys, liver and spleen, and can be fatal. This is inherited from the parent of the offspring effected through germ-line mutations, although this is a harmful condition for some places it can be beneficial. For example, in places where malaria is common, the sickle cell trait provides a survival advantage against malaria fatality over people with normal haemoglobin in regions where malaria is endemic.

Cystic fibrosis is....

6.2 Population Genetics

A **population** is a group of individuals belonging to the same species that are able to interbreed.

A **gene pool** is the combined genes of a population, including the different alleles for each gene.

Variation is created in a population by sexual reproduction and mutations.

Allele frequency is how often each allele for a gene occurs within a population and is often expressed as a percentage.

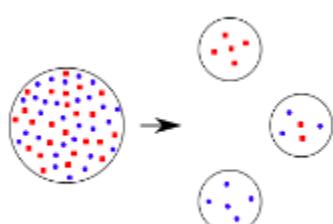
Sexual selection is a process where some traits become more common in a population due to mating partners being selected on the basis of them having those traits.

Gene flow refers to changes in allele frequency due to new individuals entering a population or from individuals exiting a population. These individuals alter the gene pool.

Genetic drift involves changes in allele frequency in the gene pool of a population due to random chance.

The **bottleneck effect** refers to when a chance event causes a drastic decrease in population size, resulting in genetic drift.

The **founder effect** occurs when a new population is started by a small number of individuals who are not representative of the original. The founder effect is the loss of genetic variation that occurs when a new population is established by a very small number of individuals from a larger population.



Allele Frequency

Calculator the frequency by:

1. Identify the amount of types each one has (how many B, how many b)
2. Plus them together and times them, and then to find one frequency divide it by the number of the frequency you're trying to find.
3. Change into percentage

SNP's

- 99.9 % of your DNA is identical to any other person
- The 0.1% variation explains the different phenotypes between individuals

SNPs

- A variation at a single base pair
- Created by a base substitution mutation
- Approximately 10 million in the human genome
- **Single nucleotide variants (SNV)** are when one base is different at a given location
- If an SNV occurs in 1% or more of the population it is called a single nucleotide polymorphism SNP

Two differences between a SNP and an allele (2d)

- An allele is a variant for a gene, so occurs in a coding region. A SNP can be in coding or non-coding sections
- An allele can involve multiple differences in base sequence, whereas a SNP is only one base substitution
- The maximum number of variants possible for any one SNP is 4 (4 base types)

What are single nucleotide polymorphisms (SNPs)?

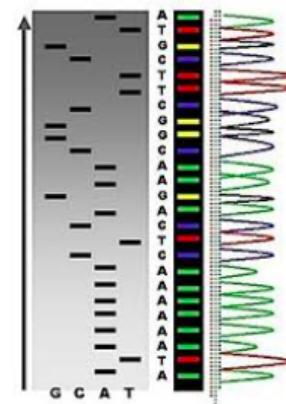
Single nucleotide polymorphisms, frequently called SNPs (pronounced “snips”), are the most common type of genetic variation among people. Each SNP represents a difference in a single DNA building block, called a nucleotide. For example, a SNP may replace the nucleotide cytosine (C) with the nucleotide thymine (T) in a certain stretch of DNA.

SNPs occur normally throughout a person's DNA. They occur almost once in every 1,000 nucleotides on average, which means there are roughly 4 to 5 million SNPs in a person's genome. These variations may be unique or occur in many individuals; scientists have found more than 100 million SNPs in populations around the world. Most commonly, these variations are found in the DNA between genes. They can act as biological markers, helping scientists locate genes that are associated with disease. When SNPs occur within a gene or in a regulatory region near a gene, they may play a more direct role in disease by affecting the gene's function.

Most SNPs have no effect on health or development. Some of these genetic differences, however, have proven to be very important in the study of human health. Researchers have found SNPs that may help predict an individual's response to certain drugs, susceptibility to environmental factors such as toxins, and risk of developing particular diseases. SNPs can also be used to track the inheritance of disease genes within families. Future studies will work to identify SNPs associated with complex diseases such as heart disease, diabetes, and cancer.

DNA sequencing

- Determining the order of nucleotide bases (A,T,C,G)
- DNA is cut into small segments
- Enzymes are used to make multiple copies of the segments
- These are fed into a machine
- DNA strands separate allowing dideoxy nucleotides to bind with the DNA
- Dideoxy nucleotides give off a colour signal that is read by the machine
- These colour signals allow the order of the nucleotide bases to be determined.



HGP

- The human genome project (HGP) involved the sequencing of the 3 billion bases of the entire human genome and identifying every human gene.
- The HGP was completed in 2003, ahead of schedule due to collaboration and advances in technologies.

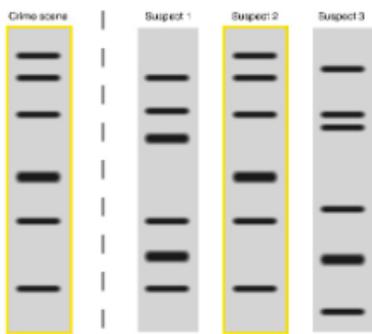
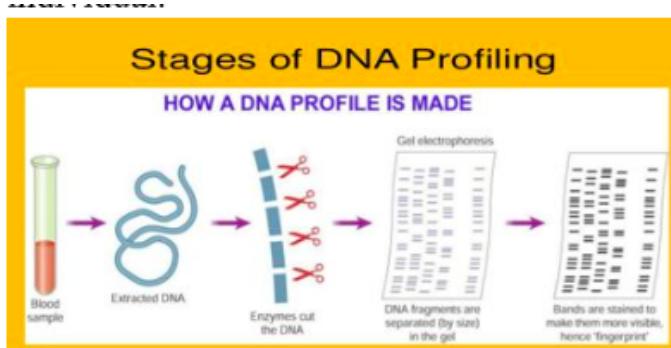
DNA profiling

Used to identify an individual and to determine if two individuals are related to each other.

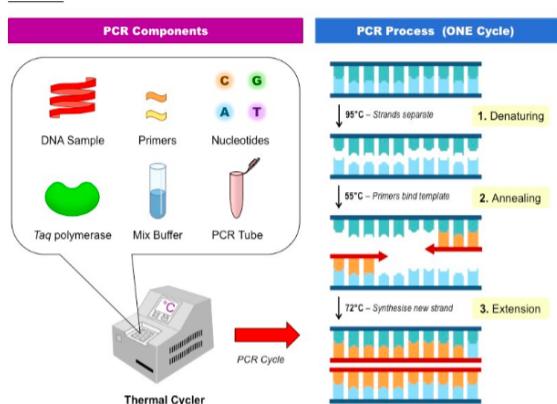
STRs – short tandem repeats (in non-coding sections of DNA) – vary in length between individuals due to different numbers of repeats.

Steps in DNA profiling

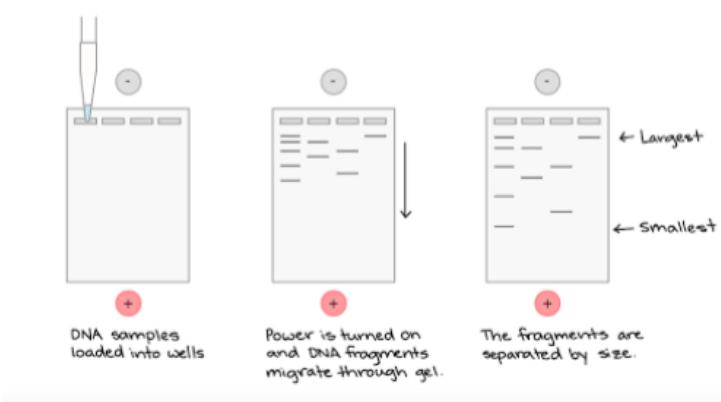
1. DNA is extracted and purified from a suitable sample, e.g. Blood, hair etc.
2. Multiple copies of STRs in the DNA are made using PCR – polymerase chain reaction.
3. Gel electrophoresis is used to separate the STRs based on their differing lengths.
4. The gel results display the length for each STR for an individual. These can be compared to other DNA samples to determine relationships or for forensic identification of an individual.



PCR:



Gel electrophoresis:



Using population data in conservation genetics

Conservation genetics – the use of data on the DNA and genes of a population to help guide management decisions that seek to preserve the population.

Used to assist the preservation of endangered species.

Genetic diversity is important for the long-term survival of a population.

DNA sampling can give an indication of genetic diversity.

Ethics

Just because you can do something, should you do it?

Is it moral or ethically ok?

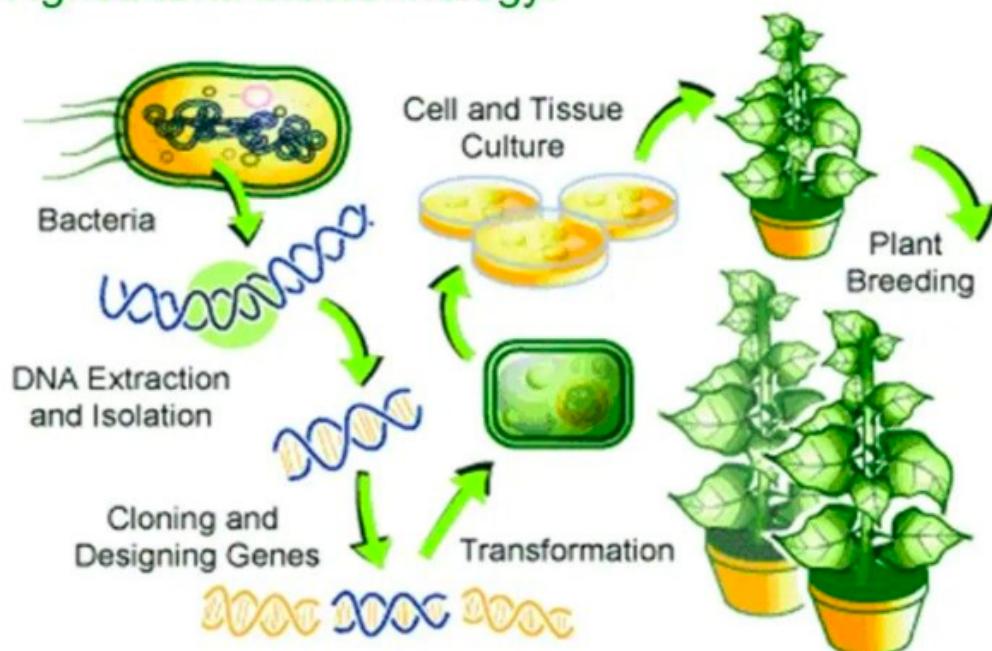
Religion, laws, culture and personal attitudes can all influence one's ethics and cause differences between each individuals' ethics

Biotechnology

Biotechnology = the manipulation of living organisms and/or parts of living organisms in a vast range of products and processes.

Modern biotechnology often uses cells as 'factories' to produce useful proteins, which can then be used in industrial applications, food or medicine.

Agricultural Biotechnology:



Module 7:

Infectious

disease

7.1 Disease and pathogens

A disease is any condition that impairs the normal functioning of an organism

Infectious disease:

Pathogens:

- is caused by a causative agent like viruses, bacteria and fungus
- it MAY be transferred from one person to another
- the transfer may be direct, from person to person, or it may be carried out by an intermediary (called a vector), such as a blood sucking insect (mosquito).
- examples of infectious disease include: colds, influenza, chicken pox, herpes, measles.

Non-infectious disease:

- are due to disease-causing organisms
- are related to lifestyle or environment, such as cardiovascular disease and skin cancer
 - genetic disorders like down syndrome
 - cellular malfunction like breast cancer

Way to remember the difference: is there a bacteria involved that has transferred the disease to the person e.g through sex, from eating bad food, coughing on someone, from an animal

Infectious disease caused by virus: influenza

Infectious disease caused by bacteria: salmonella, tetanus

Non-infectious disease caused by environment: sunburn, skin cancer

Types of Pathogens:

Pathogens are organisms or biogenic molecules (such as a protein) that cause diseases. They are infectious agents, meaning they can be passed from one organism to another.

Types of pathogens:

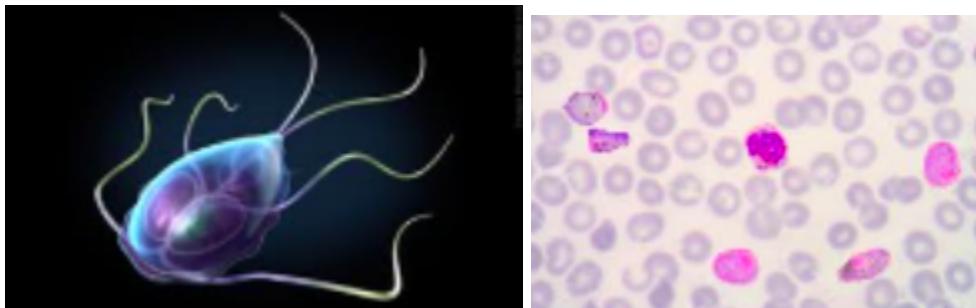
1. Macro-parasites (you can visually see the pathogen) e.g ticks, fleas, leeches, tapeworm



2. Fungi e.g Tinea pedis (a mould like fungus feeding on skin)



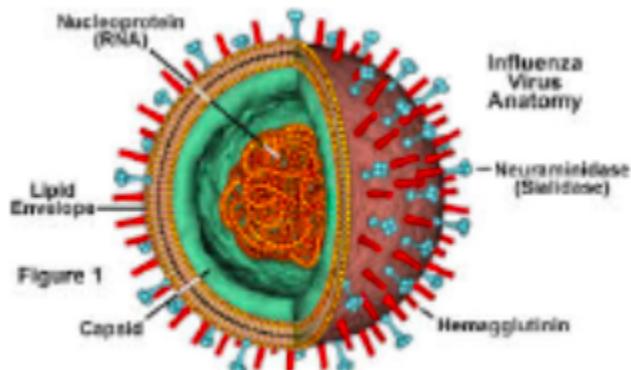
3. Protozoans - single celled eukaryotic organisms e.g malaria, giardia



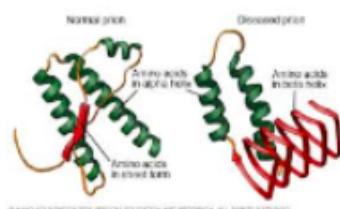
4. Bacteria - single celled prokaryotic organisms e.g clostridium tetani, salmonella



5. Viruses - small capsule of protein containing DNA or RNA e.g influenza, measles, polio, rabies, the common cold. Only way to fight it is your immune system, and get vaccinations.



6. Prions - protein molecules, which reproduce themselves and cause disease, especially of nerve tissue such as the brain. e.g BSE prions causes 'Mad Cow Disease'



Type of Pathogen:	Example:	Treatment:
Macro-parasites	arthropods Roundworms Live in human gut	Range of medications, Improve sanitation and education on it are preventative measures
Fungi	Saprophytes Parasites Hyphae makes up fungi Tinea on the feet, they thrive on warm, moist environments (that's why change rooms and bathrooms are easy to get from)	Creams, sprays, tablets help treat symptoms
Protozoa	Pathogenic protozoa Trophozoite stage Malaria Useful examples:	Treatment with antibiotics, but can become resistant. Treatment research is ongoing
Bacteria	Bacilli - rod shaped Cocci - spherical shaped (boils, gonorrhoea) Spirochaetes - tiny spiral shapes (lyme disease, syphilis) Helicobacter pylori - causing stomach ulcers Salmonella Food poisoning strep throat	Antibiotics, like penicillin
Viruses	HIV and aids Ebola Influenza Considered non-living Spreads through body through blood and lungs	Preventive methods like vaccines, help the immune system fight when infected.
Prions	Mad cow disease The immune disease does not have symptoms on the bovd, as you are born with them	No treatment

Order from smallest to largest:

Prions, viruses, bacteria, fungi, protozoa, macro-parasites

Type	Type of cell	No. of cells	Typical size	Structure
Bacteria	Prokaryotic	unicellular	1um	Prokaryotic cells with cell walls. DNA present but no nucleus. Often has structures to aid motility (movement).
Fungi	Eukaryotic	both	4um (for uni)	Eukaryotic cells with cell walls. DNA is inside a nucleus. Yeasts are often round in shape, but fungi can also form irregular branching shapes.
Protozoa	Eukaryotic	unicellular	50um	Eukaryotic cells with no cell walls. Often has structures to aid motility (movement).
Viruses	non-cellular	none	80 nm	A protein outer case protects internal nucleic acid (can be RNA or DNA)
Prions	non-cellular	none	1nm	A protein.
Macro	Eukaryotic	multicellular	> 1mm	A wide variety of multicellular eukaryotic organisms.

Transmission of disease:

epidemic = a situation when over a short period of time, many people in a region contract a specific disease

pandemic = a situation when over a relatively short period of time, many people worldwide contract a specific disease as it spreads from region of origin

Pathogens must use one of several modes of transmission between hosts to host.

This can be direct contact, indirect contact or vector transmission.



Direct transmission - this involves the individuals physically transferring the pathogen e.g through touching, kissing, sexual intercourse.

Indirect transmission - this involves objects being contaminated with pathogens e.g a used tissue, a fork with saliva traces, used bed sheets, or contaminated medical equipment (these contaminated objects are known as fomites)

Vehicle transmission - this involves the spread of pathogens by contaminated air, food or water

Vector transmission - this involves animals assisting in transfer of pathogens between individuals e.g malaria being transferred from mosquito to human.

Treatment of drinking water

Coagulation, flocculation and sedimentation.

Some particles will spontaneously settle out from water on standing. (Sedimentation)

A chemical called a coagulant is added. It reacts with the particles in the water, forming larger particles called flocs which can be filtered out.

Filtration:

Water is passed through a filter which will remove fine suspended solids and some larger microorganisms.

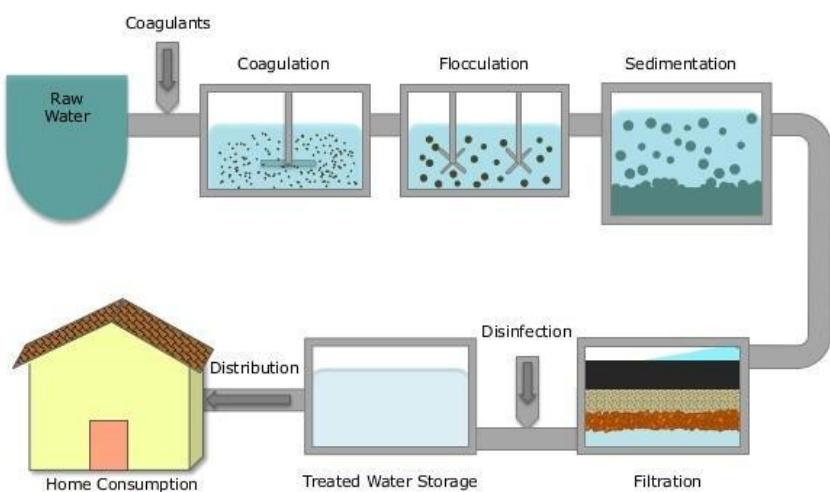
Water stabilisation

The pH of the water may need to be altered. For example, to prevent copper corrosion of pipes, many waters have lime (calcium carbonate) added.

Disinfection

Harmful microorganisms such as bacteria need to be killed either chemically or with ultraviolet radiation. Chemical disinfection includes chlorination, and ozone treatment

Water Treatment Process



Water purification experimental design

7.2 Germ theory of disease

Germ theory and spontaneous generation

Miasma theory

- Disease or illness was caused by bad air.
- Malaria means bad air.
- Ancient times and the middle ages in some countries.

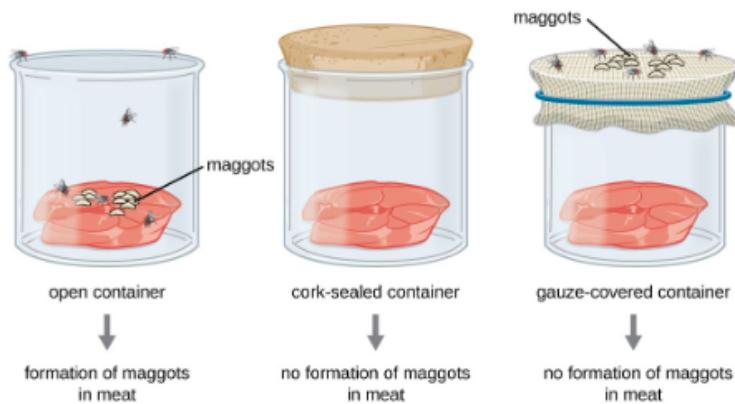
Germ theory

- Mid 1800s. Invention of the microscope allowed people to observe microorganisms.

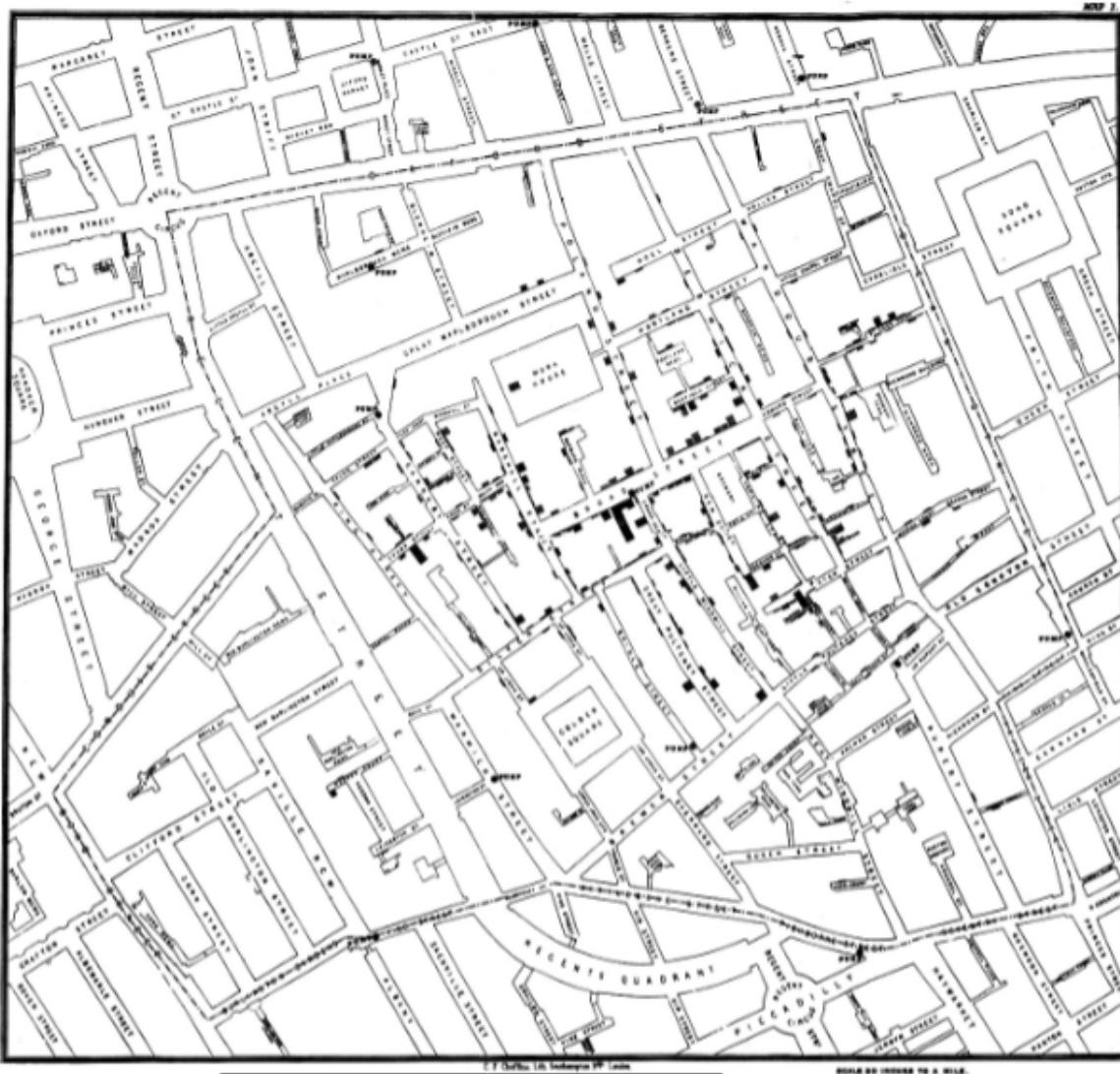
Spontaneous generation

- The process by which living organisms develop from nonliving matter.
- E.g. pieces of cheese and bread wrapped in rags and left in a dark corner, were thought to produce mice, because after several weeks there were mice in the rags.
- Many believed in spontaneous generation because it explained such occurrences as the appearance of maggots on decaying meat.

Redi's rotting meat experiment



Dr John Snow Cholera outbreak in London



Pasteurs contribution

Louis Pasteur disproved the theory of spontaneous generation and contributed to the germ theory.

He was a French biologist, microbiologist and chemist. He is credited with the discoveries of the principles of microbial fermentation and pasteurisation.

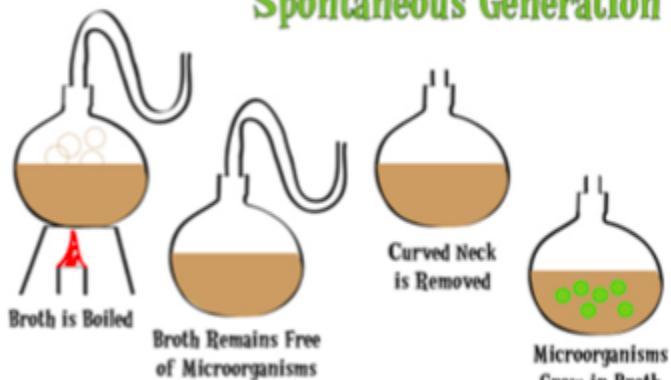
He also created the first vaccines for rabies and anthrax.

Louis Pasteur



(Fields, Date unknown)

Pasteur's Test of Spontaneous Generation



(Amoebamike, 2009)

His contribution to other diseases:

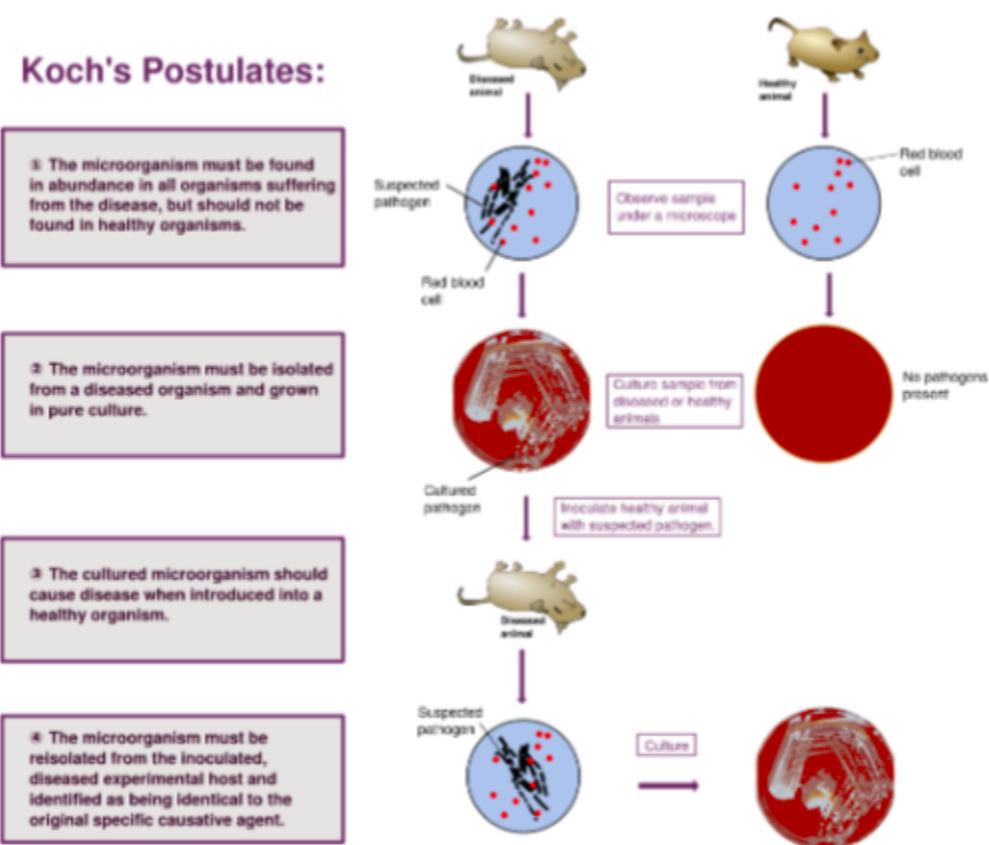
Area of work	Brief overview of Pasteur's work in this area
The silkworm industry	<ul style="list-style-type: none">Discovered microbes were the cause of a disease in silkworms that was threatening the silk industry.Developed a method to kill the microbes – thus preventing the disease and saving the silk industry.
A vaccine for chicken cholera	<ul style="list-style-type: none">Grew bacteria that caused cholera in chickens.Injected chicken with cholera bacteria cultures that had been left idle for some time.Found that these chicken could then be injected with fresh chicken cholera bacteria and not become diseased.
A vaccine for anthrax in farm animals	<ul style="list-style-type: none">Publicly demonstrated in multiple farm animals that injections of weakened anthrax bacilli protected the animals from anthrax when later injected with virulent anthrax bacilli.
A vaccine for rabies	<ul style="list-style-type: none">Could not identify the microbe that caused rabiesExperimented by injecting animals with body fluids from other animals with rabies and found he could prevent rabies developing in them.Successfully used this vaccine on humans (the initial test subjects had been bitten by rabid animals and were expected to soon die.)

Robert Koch and his postulates

Robert Koch was a German scientist who made many contributions to microbiology. He developed the agar plate technique that is still used today.

He used agar plates to isolate the anthrax bacillus and showed through his ‘postulates’ that this was the cause of the disease anthrax.

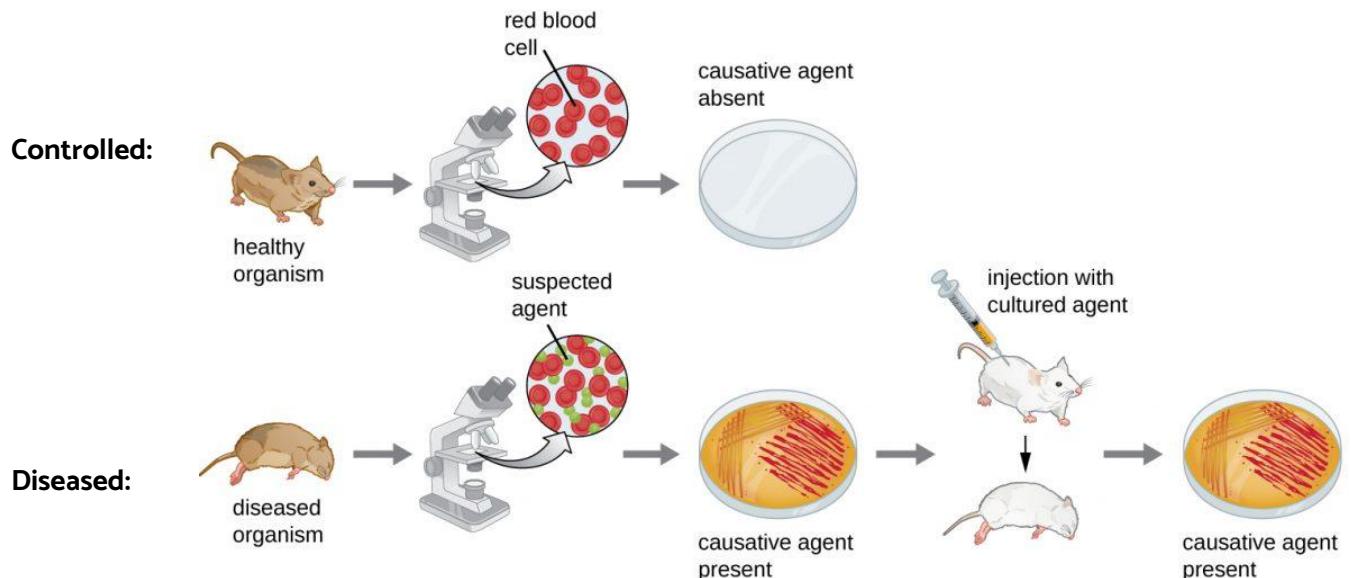
(a thing suggested or assumed as true as the basis for reasoning, discussion, or belief.)



1. The suspected causative agent must be absent from all healthy organisms present in all diseased organisms.
2. The causative agent must be isolated from the diseased organism and grown in pure culture

3. The cultured agent must cause the same disease when inoculated into a healthy susceptible organism.

4. The same causative agent must be reisolated from the inoculated, diseased organism.



Why it wont work:

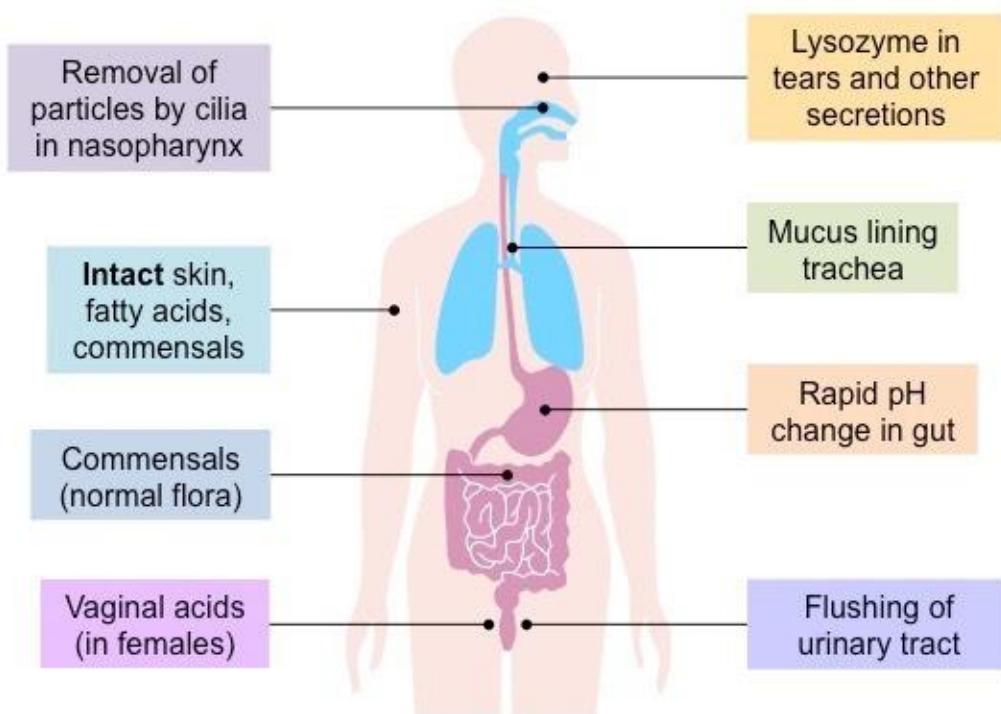
- if you can't isolate an organism in pure culture e.g viruses
- if multiple organisms cause the same disease
- if one organism leads to multiple diseases

7.3 Immune System

Lines of defence

1. Barriers to entry (non-specific)
2. Innate response (non-specific mechanisms that destroy any pathogens)
3. Adaptive immune response (specific mechanism that destroy specific pathogens)

Barriers to entry



Things that are barriers include:

- Skin:

The outside surface of skin is layers of dead, dry cells, virtually impossible for microbes to penetrate. It is a difficult environment for a pathogen to grow on (no water). Skin constantly flakes off, carrying microbes away. This provides an external barrier, strengthening keratin proteins.

- Mucous membranes

These membranes line the natural body openings of mouth and throat, and the urinary and reproductive tracts. The mucous membranes secrete mucus, a sticky fluid which traps pathogens. In some places the membranes are lined with cilia.

- Cilia

Cilia are microscopic hairs which “beat” in a rhythmic way to move mucus (and trapped pathogens) along for disposal. For example, mucus in the breathing tubes is moved upwards, until it can be swallowed into the acid of the stomach.

- Chemical barriers

Stomach is highly acidic. This kills most pathogens that are swallowed with food, or mucus. Urinary and reproductive openings are mildly acidic, enough to inhibit the growth of many microbes.

Antigens are molecules that trigger the immune response

- any substance that is usually foreign to an organism's own body = protein molecules that trigger an immune response
- viruses, bacteria, toxins and foreign proteins.
- each pathogen has its own antigen.

e.g peanuts, pollens, something that doesn't belong there.

Microflora:

These bacteria and fungi are not pathogenic. They are found on the skin, in the digestive tract and vagina, and on mucosal membranes. They reduce the ability of pathogens to access habitat and nutrients.

Innate response (non-specific mechanisms that destroy any pathogens)

Fast response that provides general protection (not antigen specific just attacks anything foreign)

The adaptive immune response involved specialised leukocytes (white blood cells) that can mount a specific response to an antigen. It involved two types of cells:

- T cells
- B cells

Individual T and B cells differ in the receptors they have on their surfaces, when it binds to an antigen it becomes activated. After primary exposure to a specific antigen, memory B and T cells are formed, circulating the body acting as a database for how to respond to specific antigens if a secondary exposure occurs.

Purpose of memory cells is that the immune system is able to recognise and respond effectively the second time.

Two types of immunity: innate and acquired

Histamines are the major part of the immune system that respond to allergic responses, humans take antihistamines to help prevent this.

Pus is formed from dead neutrophils

Type of lymphocytes:

	B cells	T cells
Where it develops	Bone marrow	Bone marrow
Where it matures	Bone marrow	Thymus gland
Type of response	Humoral	Cell-mediated

Helper T cells: Activate and direct other immune cells

Cytotoxic cells: Use chemicals to kill antigen-presenting cells

Memory T and B cells: provide future immunity

Plasma B cells: Produce many antigens-specific antibodies

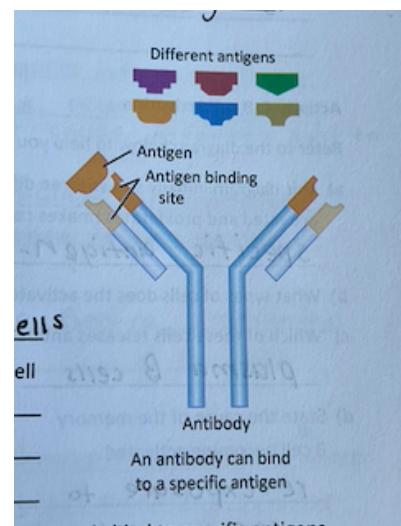
Main types of B cells:

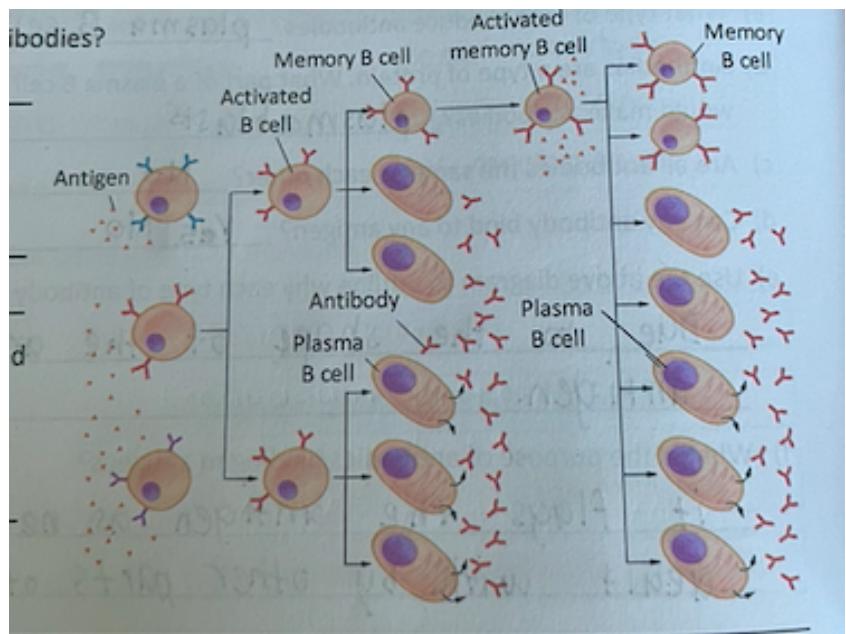
Plasma - mass produce the same antibody

Memory - helps provide future immunity

Certain antibodies can only bind with specific antigens, why? Antigen binding sites on antibodies have a specific shape or structure, which needs to match the shape or structure of the antigen

Their purpose is to help flag the antigen as needing to be dealt with by other parts of the immune system (e.g. phagocytes).





Referring to the graph:

- Only 1 antibody becomes activated because it is the only B-cell with a receptor that matches the specific antigen and so can bind to it.
- Plasma B and B memory cells activate from a normal B cell.
- Plasma B cells produce antibodies.
- Memory cells become activated when it has been re-exposure.

Types of T cells:

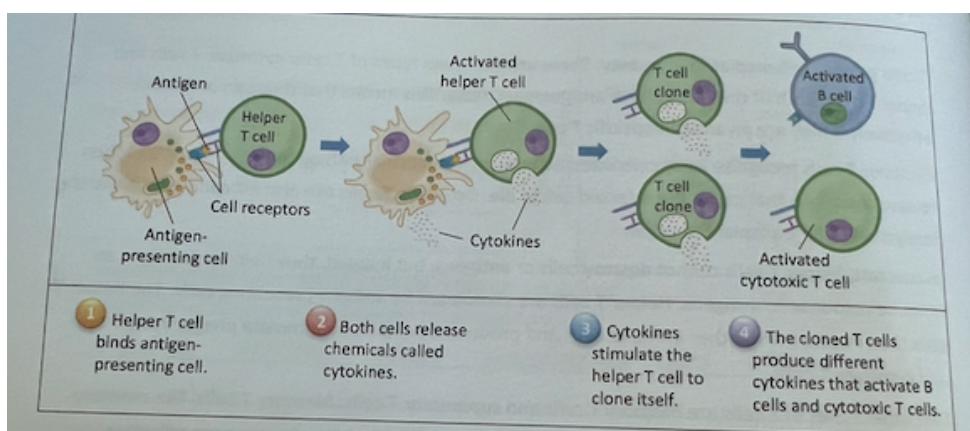
Cytotoxic - recognize infected cell e.g virus and release chemicals that cause infected cell to die

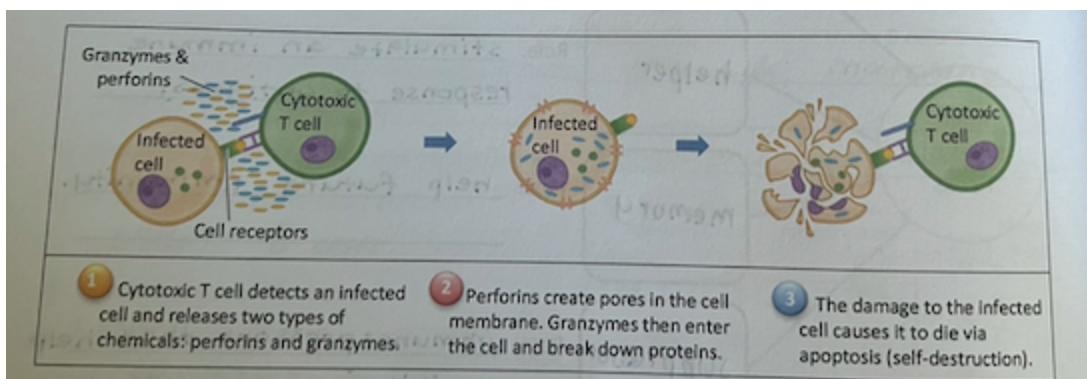
Helper - stimulates an immune response to the antigens

Memory - help future immunity

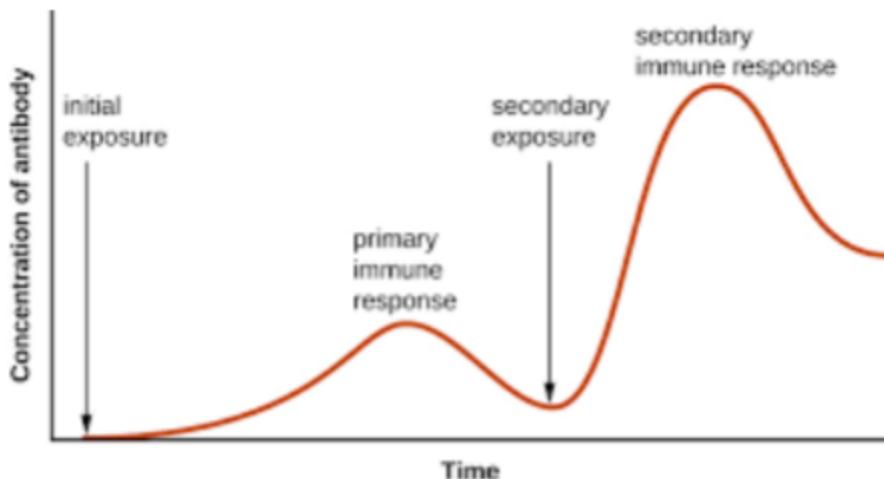
Suppressor - immunosuppressors that help inhibit immune cells at the end of an immune response.

The reason why cytotoxic T cells can combat an infection over plasma B cells is because they kill the actual infection, while B cells only kill the infection in fluid.





IMMUNE RESPONSE GRAPH



The immune response in other animals

- All vertebrates have similar immune systems, both innate and adaptive.
- White blood cells play a significant role in vertebrate immune responses.
- This similarity supports the theory of evolution, suggesting that all vertebrates evolved from a common ancestor.
- It is unlikely to have developed by chance via convergent evolution.

- Many invertebrates (no endoskeleton = no bone marrow) have hemolymph instead of blood.
- Haemolymph contain haemocytes that can ingest pathogens by phagocytosis and can form cellular capsules that surround multicellular parasites.

- Non-specific antimicrobial chemicals can assist in binding and killing pathogens.

7.4 Vaccinations

Vaccination is the administration of a vaccine to help the immune system develop protection from a disease. Vaccines contain a microorganism or virus in a weakened, live or killed state, or proteins or toxins from the organism.

Active and Passive Immunity

Naturally acquired active immunity occurs when the person is exposed to a live pathogen, develops the disease, and becomes immune as a result of the primary immune response.

Artificially acquired active immunity can be induced by a vaccine, a substance that contains the antigen. A vaccine stimulates a primary response against the antigen without causing symptoms of the disease

Naturally acquired passive immunity occurs during pregnancy, in which certain antibodies are passed from the mother's blood into the foetal bloodstream of an unborn baby. It also includes antibodies in the mother's breast milk being consumed by the baby.

Artificially acquired passive immunity is a short-term immunisation by the injection of antibodies, such as gamma globulin, that are not produced by the recipient's cells.

7.5 Antibiotics

Antibiotics treat diseases caused by bacteria

Antibiotics are chemicals which kill BACTERIA not viruses. Antibiotics are useless against "the common cold".

A "cold" is a virus. Viruses need to invade body cells to live and reproduce. To kill the virus, you need to kill the body cells.

Antibiotics are often prescribed to treat secondary bacterial infections – sore throats, ear infections etc.

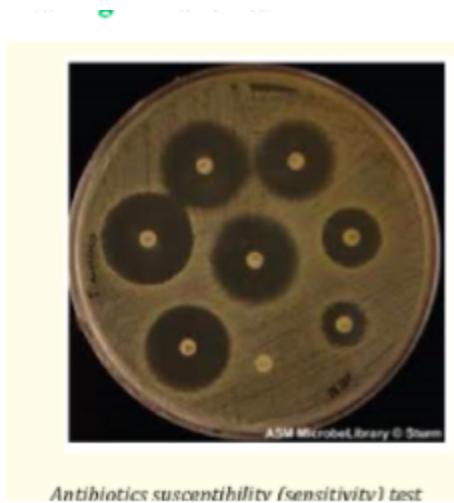
Antibiotic resistance (by natural selection)

Among the billions of individual bacteria, there may be a few which have some natural resistance to an antibiotic.

The antibiotic kills all the others, however, the resistant bacteria survive and reproduce, passing on their resistance to their offspring.

- a population of resistant bacteria.
- new antibiotics must be found

Testing antibiotics:



How do we treat diseases caused by viruses?

Viruses are intracellular pathogens. They replicate inside cells.

It is difficult to target viruses without harming the host cells

- **Antiviral drugs** exist for some viral diseases, for example, HIV (human immunodeficiency virus) and influenza, however they are not generally used for less serious diseases because of the side effects. (Nausea, vomiting, diarrhoea, nose bleeds, dizziness, headache)

This medication works by stopping the flu virus from growing.

It is used to treat symptoms caused by the flu virus. It helps make the symptoms (such as stuffy nose, cough, sore throat, fever/chills, aches, tiredness) less severe and shortens the recovery time by 1-2 days

Vaccines can be used to treat people after exposure to a virus in some cases.

These vaccines are used on viruses that take a long time to progress. They bring about an immune response in the infected person.

Traditional disease remedies

7.6 Epidemiology

EPIDEMIOLOGY is the application of scientific methods to the study of disease in populations for the purpose of prevention and control of disease. In an epidemiological study of a particular disease, a number of different aspects may be considered. These include:

- A description, or definition, of the disease.
- Possible causes of the disease.
- Occurrence and transmission of the disease
- In the population
- In the environment
- Consideration of risk factors for various members in a population, that is, who is more likely to be affected by the disease.
- Control of the disease, for example, whether an affected individual should be isolated from others.
- Prevention of the disease, for example, whether a special diet might prevent expression of the disease.
- Possible elimination of the disease.

Epidemic = a situation when over a short period of time, many people in a region contract a specific disease

Pandemic = worldwide

Endemic = the typical rate at which a disease occurs.

Prevalence = the total number of cases at a specific time.

Incidence = the number of new cases at a specific time.

Outbreak = similar to epidemic except usually refers to a smaller geographical area.

Measles in humans:

Measles in humans	
Type of pathogen	Virus
Method of transmission	Droplet infection (direct or indirect)
Typical symptoms	High fever, runny nose, red watery eyes
Possible complications	Ear infections, pneumonia, diarrhoea
Treatment	No specific antivirals kill it
Disease prevention	Vaccination - usually given as part of mumps and chickenpox

7.7 Plant defence mechanisms

- Tough lignin in bark- barrier to microbes
- Thorns, waxy cuticle
- Immune system in each cell – cell death, thickening of cell walls
- Hormonal communication to warn other parts of the plant
- Specific chemicals released

PLANT PATHOGENS



Black spot



leaf miners



Leaf curl



Mealy bug



Aphids



Leaf hoppers



Gall



Gall



Fungus

Late Blight in potatoes

Late blight in potatoes	
Type of pathogen	Phytophthora Infestans → fungi
Method of transmission	Spores that are released every 3-5 days, spread via wind and soil to new hosts and are favoured in cool and humid weather conditions.
Typical symptoms	Dark blotches on stem and leaves rots both potato and tomato plants.
Treatment	Fungicides
Disease prevention	Spray plants prior to exposure and getting fields regularly inspected.
Disease control measures	When an infected plant is found, kill all surrounding plants to stop the spread altogether.

Musa bananas

Musa bananas	
Type of pathogen	Banana bunchy top virus (BBTV)
Method of transmission	from plant to plant via aphids
Typical symptoms	<ul style="list-style-type: none">- stunted leaf growth, making leaves appear yellow and 'bunched'- prevents plant from producing fruit- affects phloem tissue meaning infected cells die a lighter colour causing the leaf stem to appear streaky.
Treatment	chemicals
Disease prevention	<ul style="list-style-type: none">- using chemical treatments- monitoring alternate vector feeding sites- removing/destroying infected plants
Disease control measures	Preventing the transportation of the infected fruit and plant material

Module 8:

Non-infectious

disease and

disorders

8.1 Homeostasis

Homeostasis = the process of maintaining a stable, internal environment.

This involves temperature regulation, pH, water and salt balance. Oxygen and carbon dioxide levels.

Blood sugar levels etc.

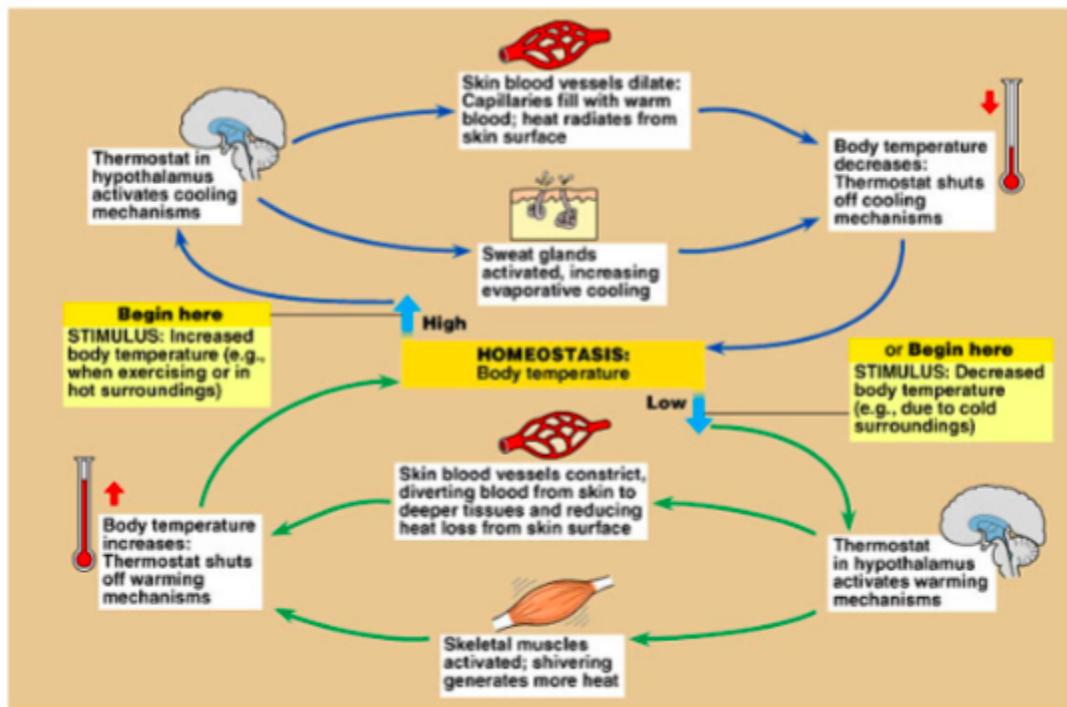
Feedback mechanisms = A situation where the result of some action feeds back into the system to control the next change to the system.

Homeostasis is always a negative feedback mechanism. The change causes the next change to be in the opposite direction e.g. as body temperature rises the mechanisms cause the body to cool down.

Homeostasis consists of two stages:

- **Detecting changes from the stable state.** receptors detect a changes (stimuli) and send message to CNS (brain/spinal cord)
- **Counteracting changes from the stable state.** CNS determines response, sends message to effectors e.g. Muscles and glands → response

TEMPERATURE FLOWCHART:



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Stimulus-response pathway	Functioning of the nervous system
stimulus	Any change in the environment that can be detected by receptors and triggers a response (e.g. extreme heat) is termed a stimulus.
receptor	Thermoreceptors in the skin and in the hypothalamus of the brain detect the stimulus (change in temperature) and convert this into a 'message' in the form of nerve impulses, which travel along nerves towards the central nervous system (brain and spinal cord).
control centre	The central nervous system (CNS), made up of the brain and spinal cord, processes the information about the change in external environment. Information from particular receptors is interpreted in specific parts of the brain. The hypothalamus processes information about body temperature (as well as hunger and thirst). Connects the receptors with the effectors.
messenger	Motor nerves carry information (as nerve impulses) from the CNS to the effectors.
effector	Muscles or glands receive impulses (via motor nerves) from the CNS; these impulses instruct the effectors to bring about a response—for example, sweat glands produce sweat to cool the body.

Endotherms = organisms that can maintain a constant body temperature by metabolic activity (warm blooded – don't use this term)

Ectotherms = organisms use the external environment to maintain body temperature. (cold blooded – don't use this term)

Adaptations

An adaptation is - any alteration in the structure or function of an organism or any of its parts that results from natural selection and by which the organism becomes better fitted to survive and multiply in its environment.

- the ability of a species to survive in a particular ecological niche, especially because of alterations of form or behaviour brought about through natural selection.

Structural adaptations are special body parts of an organism that help it to survive in its natural habitat. These changes do not occur during an organism's lifetime; rather, they accumulate over many generations.

e.g Duck has webbed feet to enable it to move faster on water to catch prey or avoid predators

Physiological adaptations a metabolic or physiologic adjustment within the cell, or tissues, of an organism in response to an environmental stimulus resulting in the improved ability of that organism to cope with its changing environment.

e.g Camels produce concentrated urine compared to other mammals as it has to reduce the water loss, this is a kind of physiological adaptation as the kidneys of camels are designed in such a way that the wastage of water is minimum.

Behavioural adaptations include activities that help an animal survive. Behavioural adaptations can be learned or instinctive (born with).

Social behaviour – some animals live by themselves, while others live in groups.

Behaviour for protection – An animal's behaviour sometimes helps to protect the animal.

e.g a rabbit freezes when it has been seen.

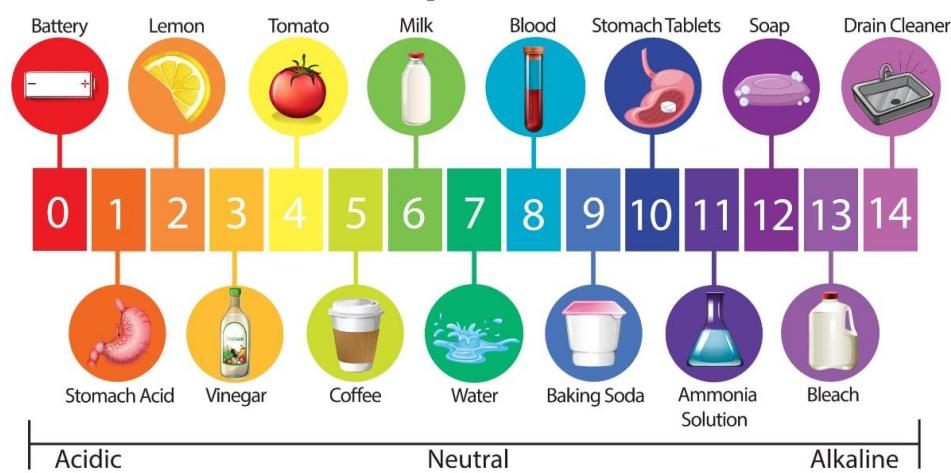
Excretion and homeostasis

Excretion is the removal of metabolic wastes and excess materials from cells.

e.g CO_2 is a waste product of cellular respiration and when dissolved in water is acidic.

A buildup of CO_2 in the blood lowers the pH of the blood (from neutral to acidic).

The pH Scale

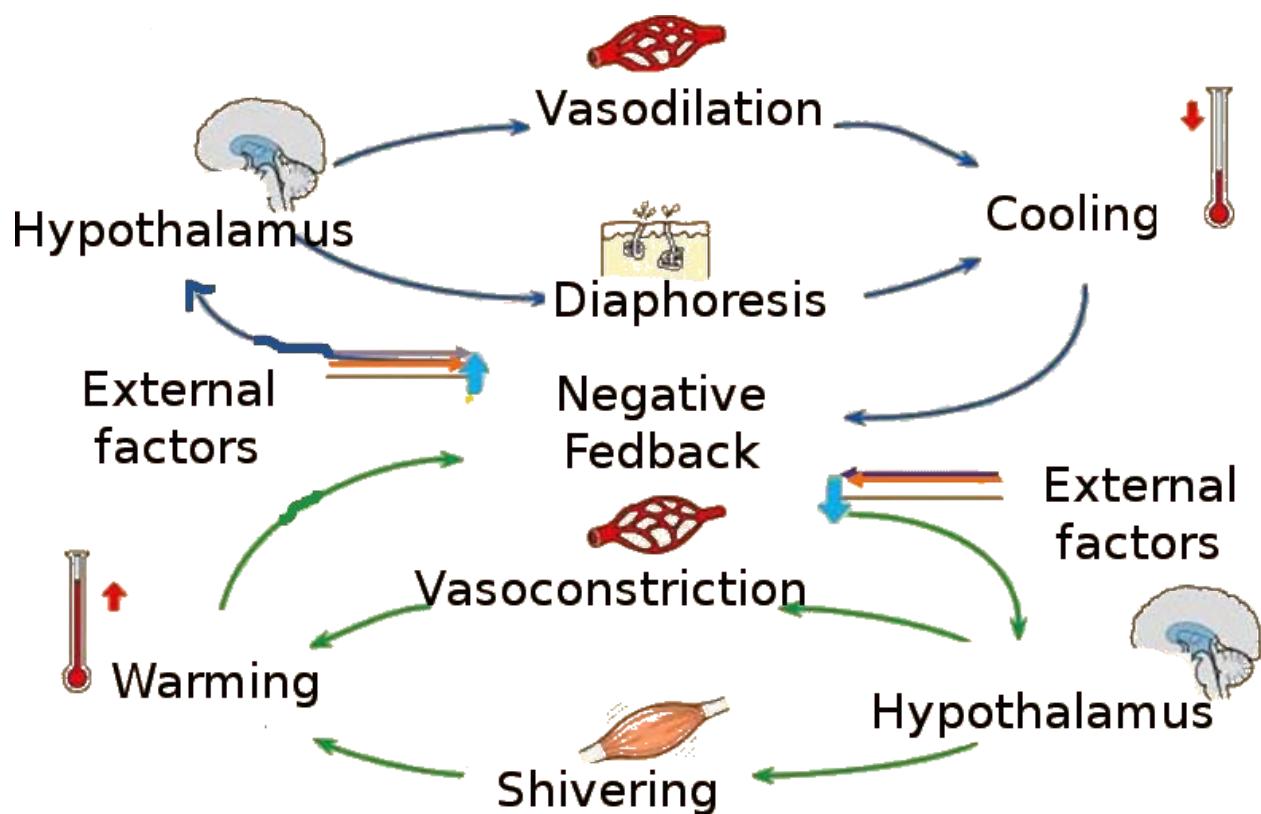


Nitrogenous wastes

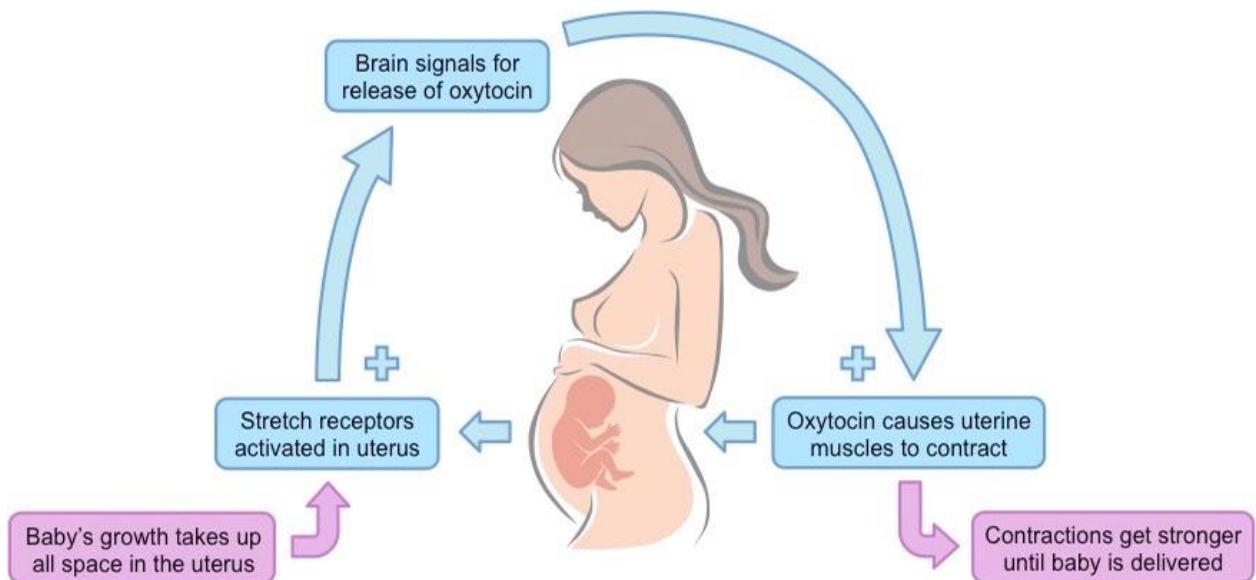
- Nitrogenous wastes are the by-product of protein metabolism.
- When proteins are broken down ammonia is formed. Ammonia is highly toxic.
- Mammals, adult amphibians and some marine organisms convert this to urea which is far less toxic.
- Insects, birds and many reptiles convert ammonia to uric acid which is also less toxic and its removal involves less water.

Fish, invertebrates and other marine organisms excrete ammonia. This is adequate because they live in an aquatic environment and the ammonia does not build up in their bodies.

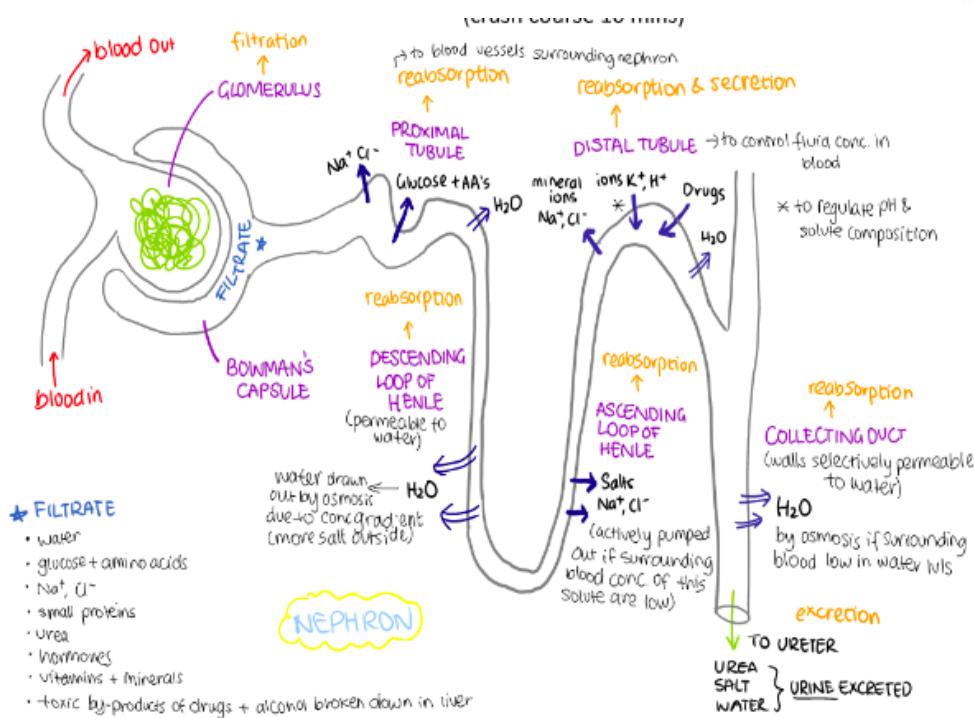
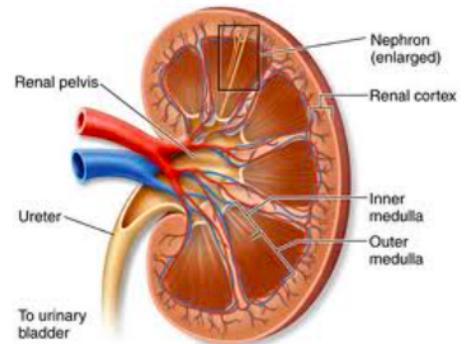
Negative feedback loop for temperature:



Positive feedback loop (only for pregnancy):



How do kidneys work?



Discussion

- Renal artery distinguished from the renal vein as it had a thicker muscular wall
- Bowman's capsule found in the renal cortex
- Cortex - darker brown colour and more striated
- Medulla - striped pattern and more red

Function of:

- Layer fat around kidney - protection and insulation
- Renal artery - carries oxygenated blood into the kidney and brings in waste ie urea
- Renal vein - carries deoxygenated blood away from kidney - less waste
- Ureter - carries urine from kidney to bladder
- Kidney tubules - reabsorption, secretion

Dialysis:

What is it? Dialysis is a procedure to remove waste products and excess fluid from the blood when the kidneys stop working properly. It often involves diverting blood to a machine to be cleaned.

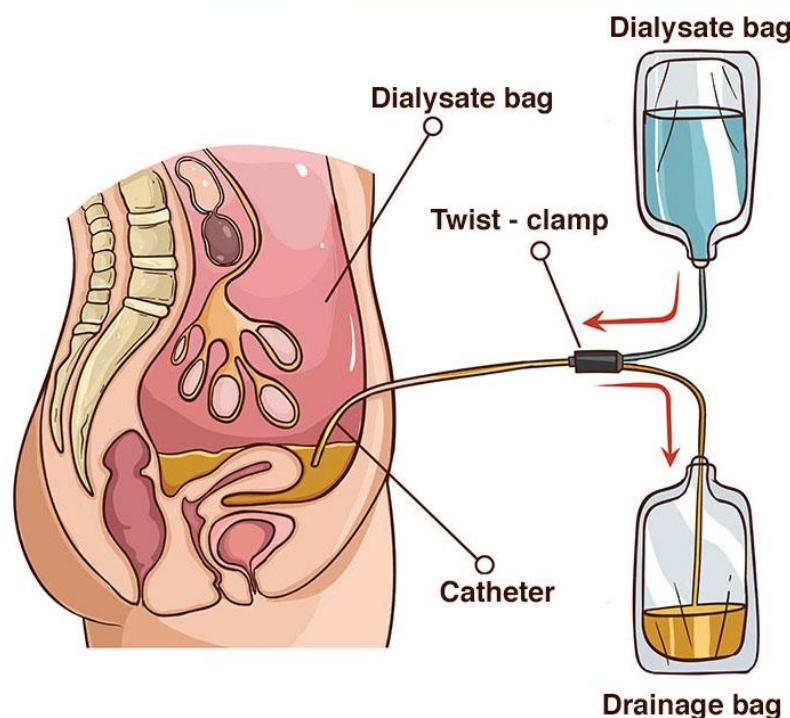
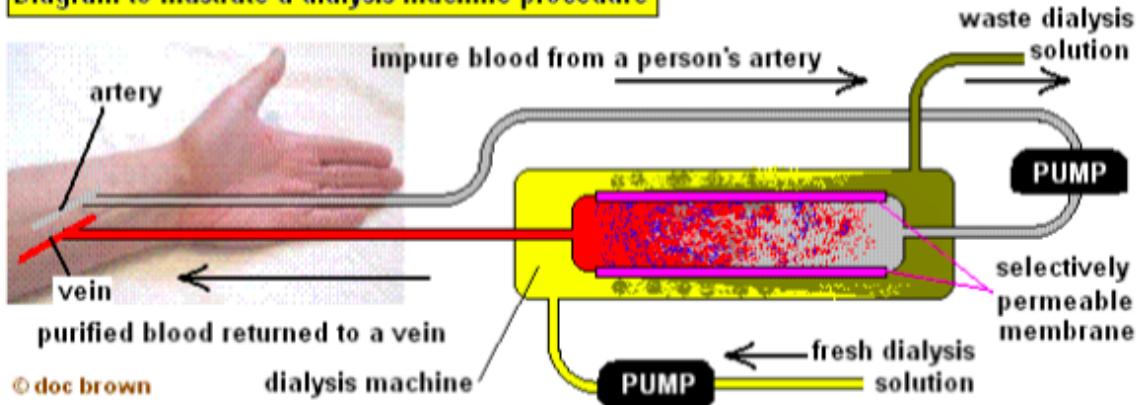


Diagram to illustrate a dialysis machine procedure



Dialysis compared to kidney:

FEATURE	KIDNEY	RENAL DIALYSIS
STRUCTURE	Consists of about 1 million nephrons, renal pelvis, and ureter.	(See diagram)
FUNCTION AND NITROGENOUS WASTES	Removes urea from blood	Removes urea from blood
OTHER FUNCTIONS	Maintains body's balance of various salts, e.g. sodium, potassium, calcium, phosphate. Hormones released into the bloodstream that regulate blood pressure and osmoregulation.	Concentrations of desired solutes can be adjusted by altering the composition of the dialysis fluid to maintain natural concentration for healthy blood.
HOW OFTEN IT OCCURS	Continuously. Approx. 1.5-2.5 litres of urine a day.	Haemodialysis, 3-4 hour sessions in hospital approx. 3 times a week. Peritoneal dialysis needs to be performed every day.
FILTRATION AND REABSORPTION	Filtration and reabsorption	Filtration only

8.2 Causes of non-infectious disease

1. Genetic diseases
2. Diseases caused by environmental exposure
3. Nutritional diseases
4. Cancer

Note – the causes of non-infectious diseases cannot always be neatly categorised.

E.g. Many cancers have been found to be caused by environmental factors and genetics. Some people have a genetic predisposition to developing bowel cancer, skin cancer etc.

1. Genetic disease case study - PKU
2. Environmental exposure - Lung cancer
3. Nutritional disease
4. Cancer - cervical cancer and prostate cancer

Type of exposure	Specific example	Effects
Extreme forces	A car crash	Physical injuries and trauma
Harmful chemicals	Frequent binge drinking	Reduced mental capacity and liver damage
Extreme temperatures	Being lost in snow	Tissue damage known as frostbite
Excessive noise	Balloon bursting in ear	Burst eardrum and hearing loss
High levels of radiation	Nuclear bomb	Mutations to DNA within cells
Harmful plants or animals	Touching a rose thorn	Pain and superficial skin damage

Nutritional disease:

Scurvy	
Cause	Low vitamin C, not eating enough fruits and vegetables
Symptoms	Bruising, bleeding of gums, rashes, coiled hair, weight loss, fatigue
Treatment	Medications and increase a diet rich in fruit and vegetables

Incidence - the number of new cases diagnosed

Prevalence - the total number of cases in the population

Mortality - the number of deaths due to a condition

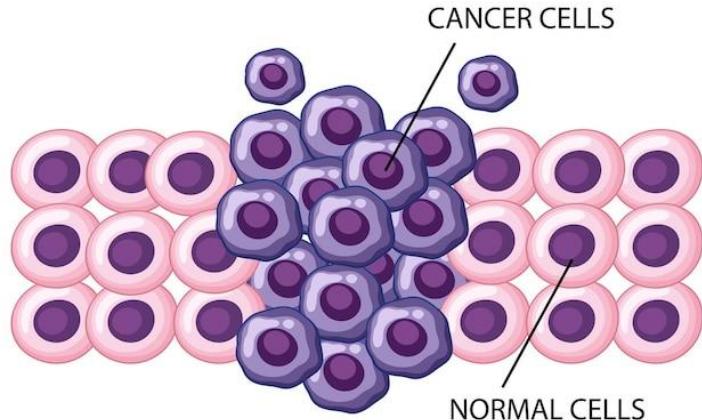
Cancerous cells

Are a result of mutations to essential genes that control growth and repair by cell division (mitosis). Due to internal or external factors.

External factors: UV rays, smoking

Internal: heredity, hormone growth

Treatments include chemotherapy, radiation and surgery.



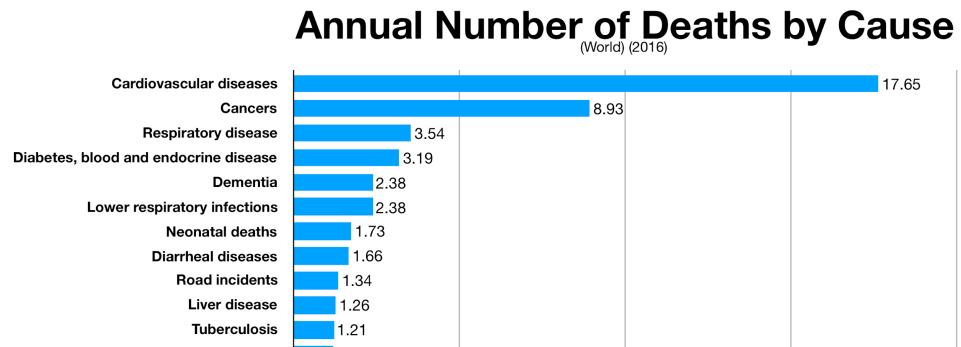
Tumour suppressor genes: help prevent cancer, they trigger cell death when cancer cells are detected.

For example the gene can suppress the BRCA1 cell that individuals can be born with.

Cervical cancer

HPV causes 99% of cervical cancer cases, as well as other sex organ cancers e.g vaginal. Most strains are asymptomatic, whilst some other strains can cause genital warts, persistent infections and development of cancer cells

Leading cause of death



More people die from non-infectious diseases, which are harder to treat and prevent, and usually cost more money for governments to implement strategies.

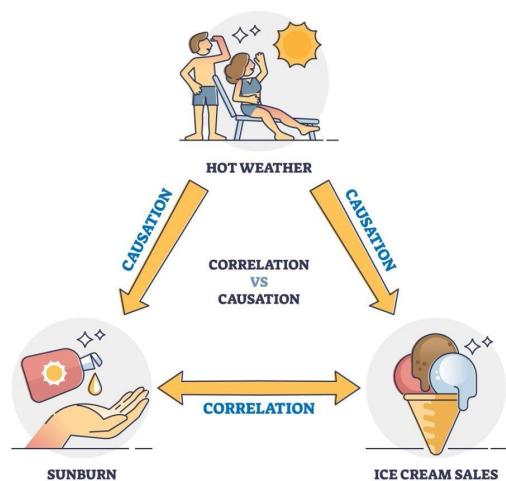
8.3 Epidemiology

What is epidemiology? The branch of medicine which deals with the incidence, distribution and possible control of diseases and other factors relating to health.

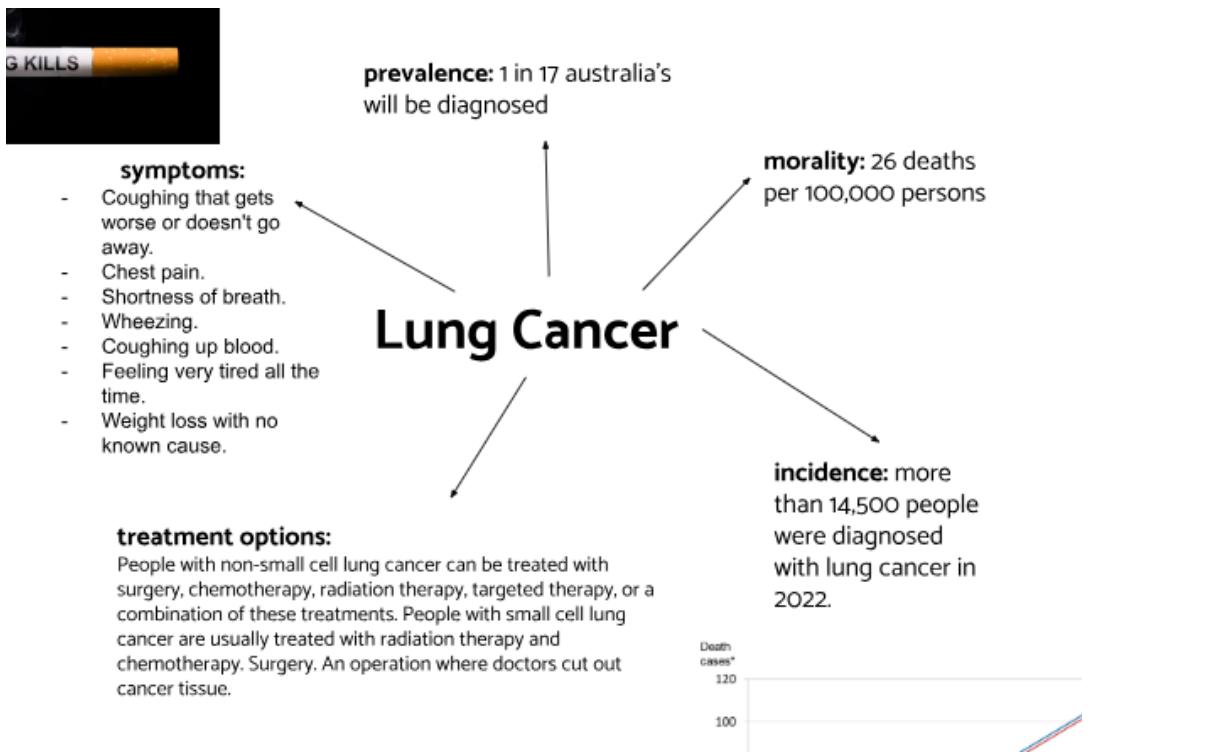
Correlation vs causation

A correlation between variables, however, does not automatically mean that the change in one variable is the cause of the change in the values of the other variable. Causation indicates that one event is the result of the occurrence of the other event; i.e. there is a causal relationship between the two events.

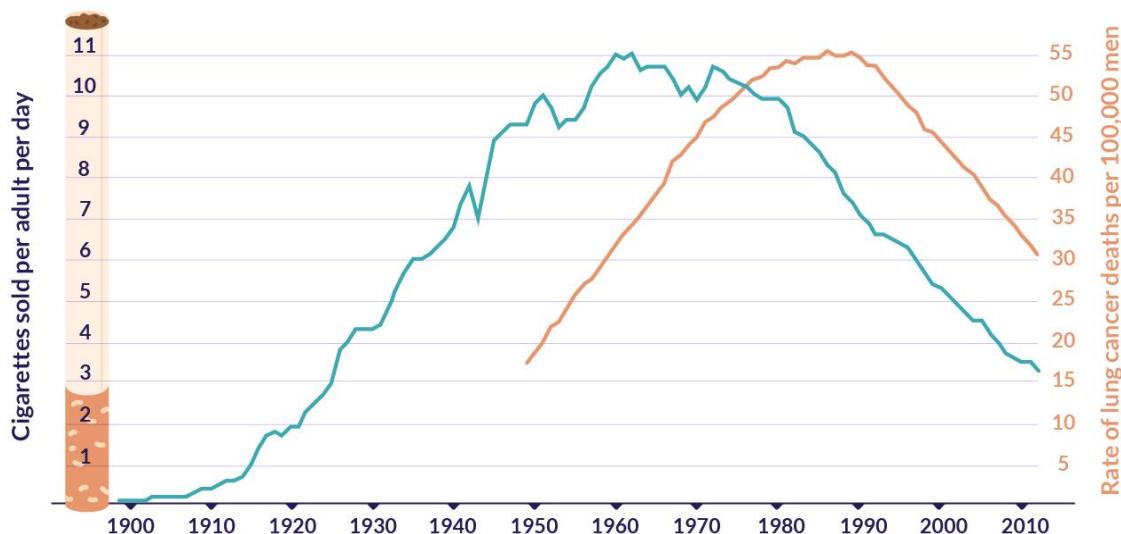
Correlation does not imply causation.



Smoking and public health:



Decline in cigarette sales and lung cancer mortality over time



Epidemiology data for diabetes:

Diabetes is one of the top 10 leading causes of death in Australia, Australia having higher prevalence.

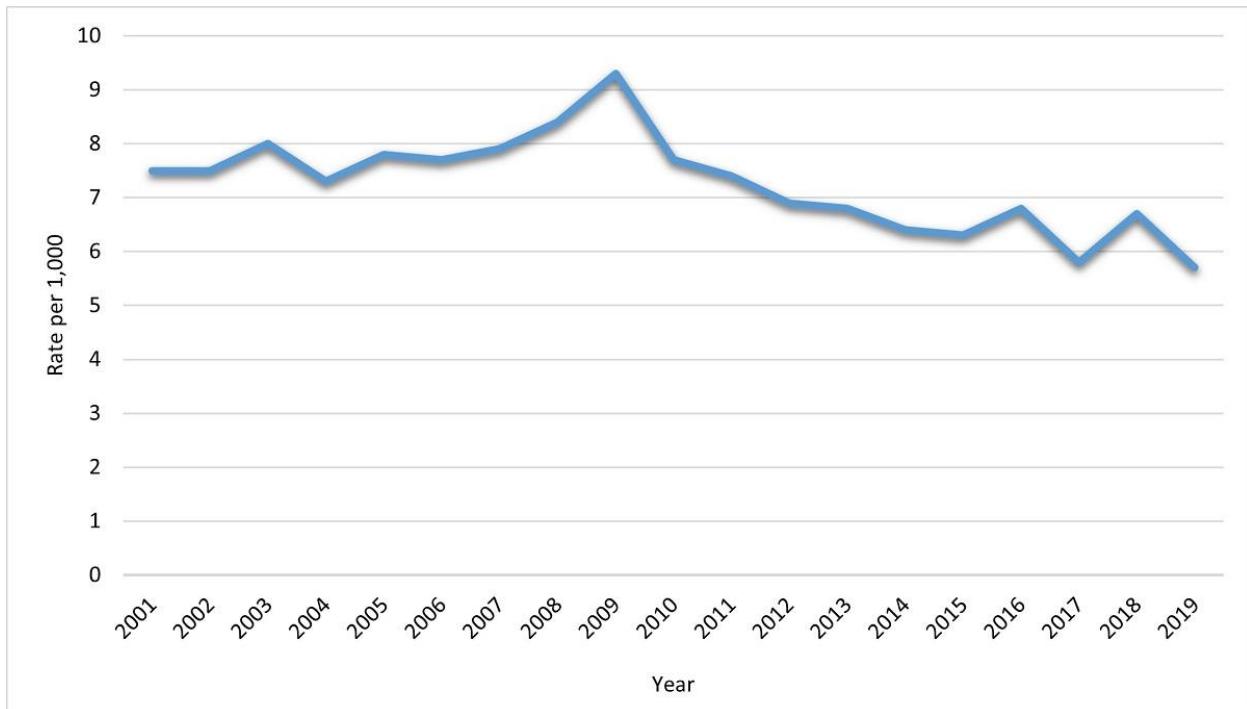
Diabetes refers to several different conditions that all involve problems maintaining homeostasis of blood glucose levels, there are three main types:

- Type 1: an autoimmune response that damages the cells of the pancreas that are involved in insulin production. Typically diagnosed during early years and childhood, managed with ongoing insulin treatment. Exact cause is unknown.
- Type 2: most common form of diabetes. It involves the pancreas failing to produce adequate levels of insulin. Various lifestyle factors contribute to it like eating unhealthy with high levels of fat and not exercising regularly (becoming overweight). In most cases, it can initially be managed through lifestyle changes, however as the condition progresses medications like insulin will be used simultaneously.
- Gestational: this occurs during pregnancy, with 13% of women in Australia becoming diagnosed with it. It usually goes away after pregnancy but increases risk of developing type 2 later in life, it is treated mostly through diet and exercise, but may also require insulin.

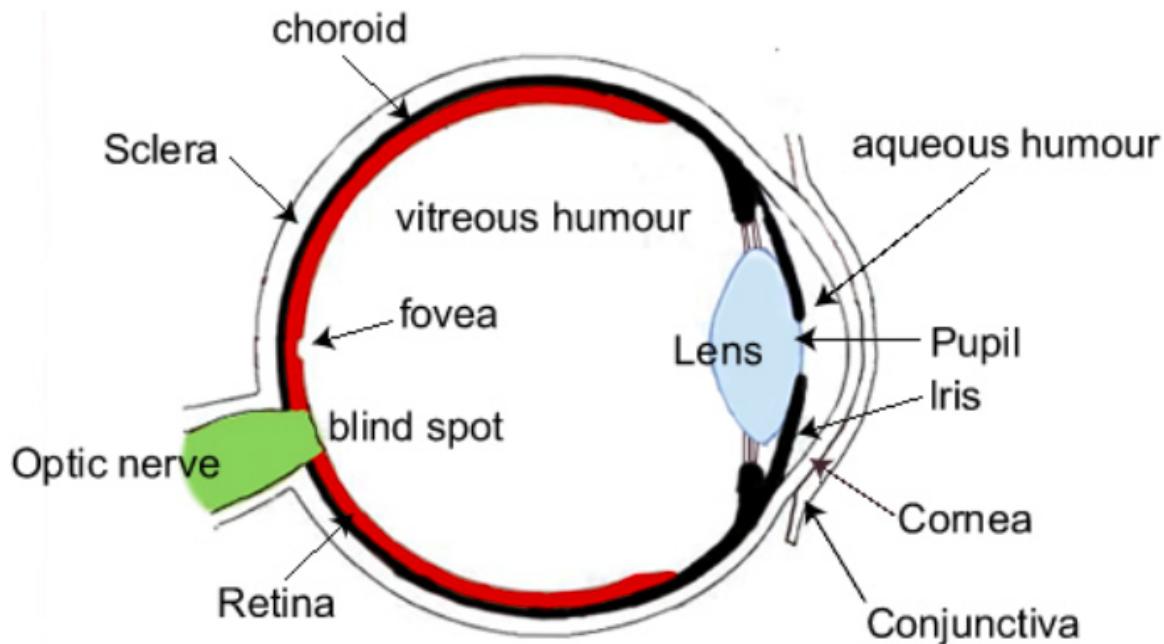
DIABETES MELLITUS

TYPE 1 VS TYPE 2

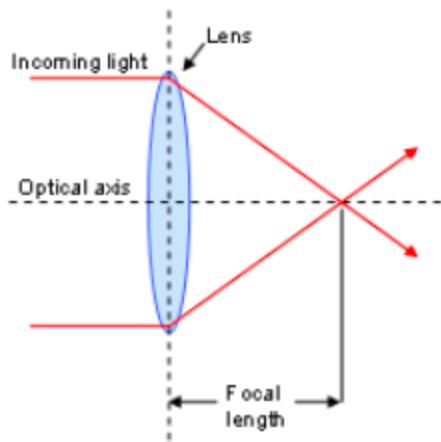
TYPE 1 DIABETES	TYPE 2 DIABETES
<ul style="list-style-type: none">• Occurs when the pancreas is unable to produce enough insulin• Tends to develop at a young age• Cannot be prevented• Require insulin therapy	<ul style="list-style-type: none">• Occurs due to insulin resistance (i.e. when the body does not respond well to insulin)• Tends to develop at an older age• Can be prevented with lifestyle changes• Can be managed with lifestyle modifications alone if diagnosed early
<ul style="list-style-type: none">• Both share symptoms of frequent urination, increased thirst, extreme hunger, unintentional weight loss, fatigue, blurry vision, sores or wounds that heal slowly, and numbness and tingling sensation in hands and feet.• Both can benefit from lifestyle modifications such as a healthy diet, physical activity, blood sugar level monitoring, and management of stress and other existing health conditions.	



8.4 Human eyes



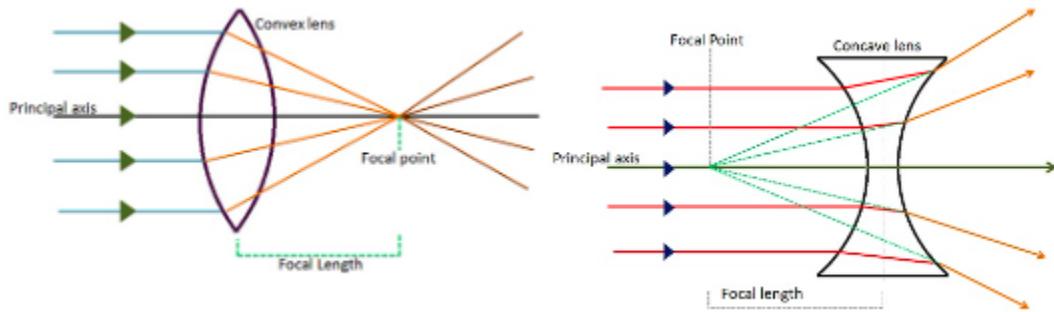
Name	Observations	Structure related to function
Cornea	Transparent and strong	Allows light through and protects t r
Sclera	Whitish and very tough	Protects eyeball
Choroid	Black	Absorbs light to prevent internal reflection
Pupil	An opening	Controlled by iris, allows light through t
Retina	Thin layer	Detects light
Iris	Rings of muscle	Controls the size of the pupil opening
Lens	Transparent, magnifying	Refracts light and allows light through t r
Aqueous humor	Transparent and watery	Allows light through and maintains shape of eye t r
Vitreous humor	Transparent and jelly-like	Allows light through and maintains shape of eye t r
Ciliary body	Ring of muscles	Changes shape of lens to focus light on the retina
Optic nerve	White, tough, tube-like	Sends electrical messages to the brain to form an image



How an image is focused on the retina and sent to the brain

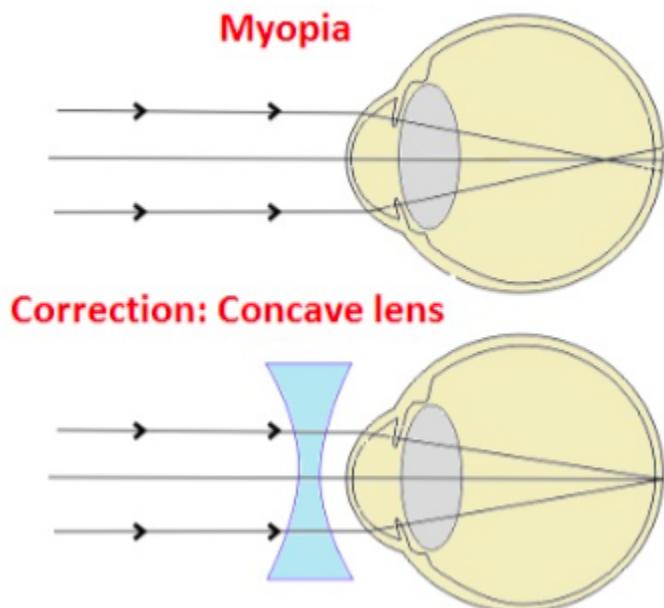
1. Light Enters the Eye: When you look at an object, light from that object enters your eye through the cornea, which is the clear front surface of the eye.
2. Refraction by the Cornea: The cornea bends or refracts the incoming light. It's curved like the front of a camera lens, and this bending of light helps to focus it.
3. Further Refraction by the Lens: After passing through the cornea, the light continues its journey through the eyes lens. The lens can change its shape, which allows it to fine-tune the focus, especially for objects at different distances.
4. Focused Image on the Retina: The combined refraction by the cornea and lens causes the light to converge and form a sharp, focused image on the retina at the back of the eye. The retina contains light-sensitive cells called photoreceptors (rods and cones) that convert this image into electrical signals.
5. Transmission to the Brain: These electrical signals are then sent to the brain via the optic nerve, where they are interpreted as the visual information that you perceive as an image.

Lens types:



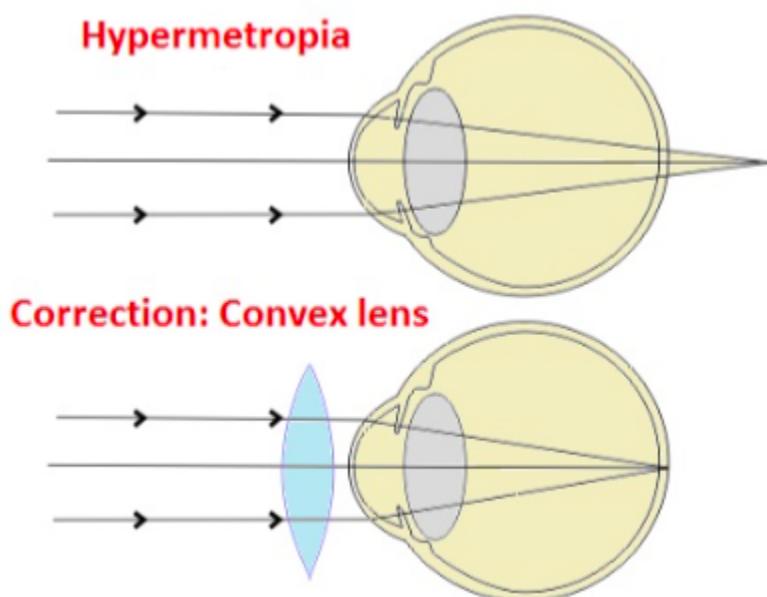
Myopia

Distant objects are not seen clearly, because the light focuses in front of the retina.



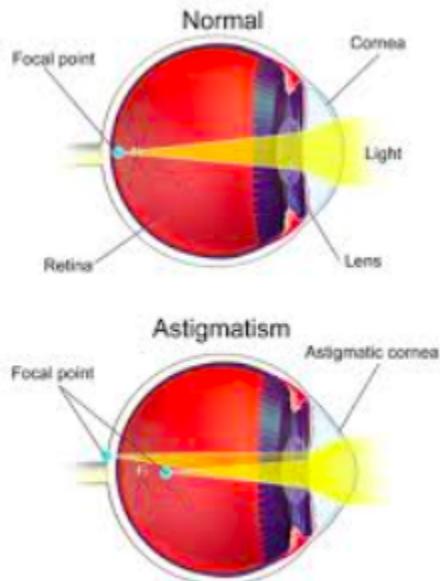
Hyperopia

Objects that are close up appear blurry because the light focuses behind the retina.



Astigmatism

The curve of the lens or cornea is asymmetrical, causing light to focus in two places, causing blurry vision.



Astigmatic cornea distorts the focal point of light in front of and/or behind the retina

Presbyopia

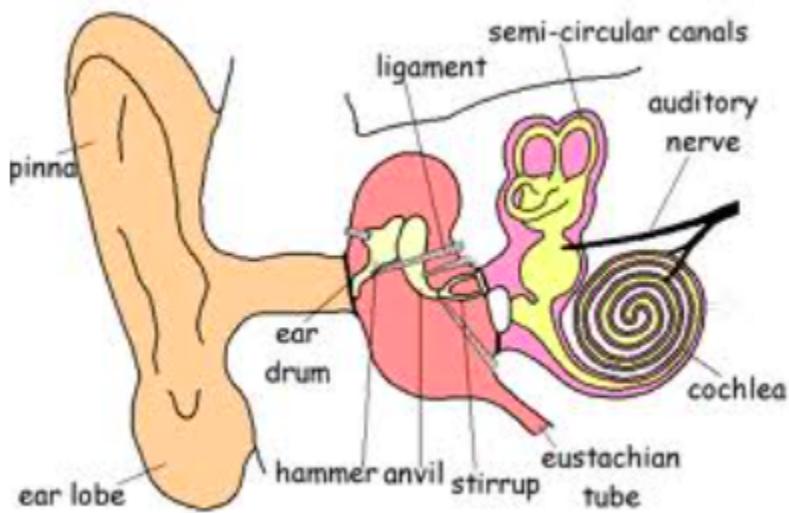
Similar to hyperopia. As people age the lens gets stiffer and less flexible making it harder for the muscles to pull the lens into shape so people can experience blurry vision of both near and far objects. Most people experience blurry vision of close up objects. The light is being focused behind the retina.

Accommodation

To see far objects, the ciliary muscles relax to cause the lens to become thinner.

To see close objects, the ciliary muscles contract to cause the lens to become rounder.

8.5 Human Ears



1. Pinna collects sound waves which pass through the ear canal.
2. Waves reach the eardrum causing it to vibrate.
3. Vibrations then pass to the middle ear (ossicles).
4. Ossicles amplify the noise and pass it to the inner ear.
5. Vibrations pass through the fluid of cochlea, making tiny hairs move.
6. This causes electrical signals to travel through the auditory nerve, to your brain.

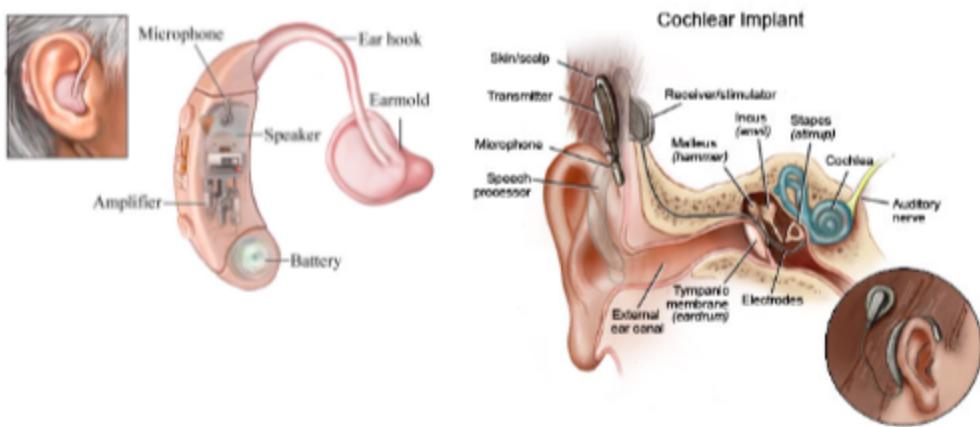
How the ears control balance

When you move your head, the fluid within the semicircular canals (which sit at right angles to each other) also moves. This fluid motion is detected by the hair cells, which then send nerve impulses about the position of your head and body to the brain to allow you to maintain your balance.

Long response:

COMPARING AND EVALUATING HEARING AIDS AND COCHLEAR IMPLANTS

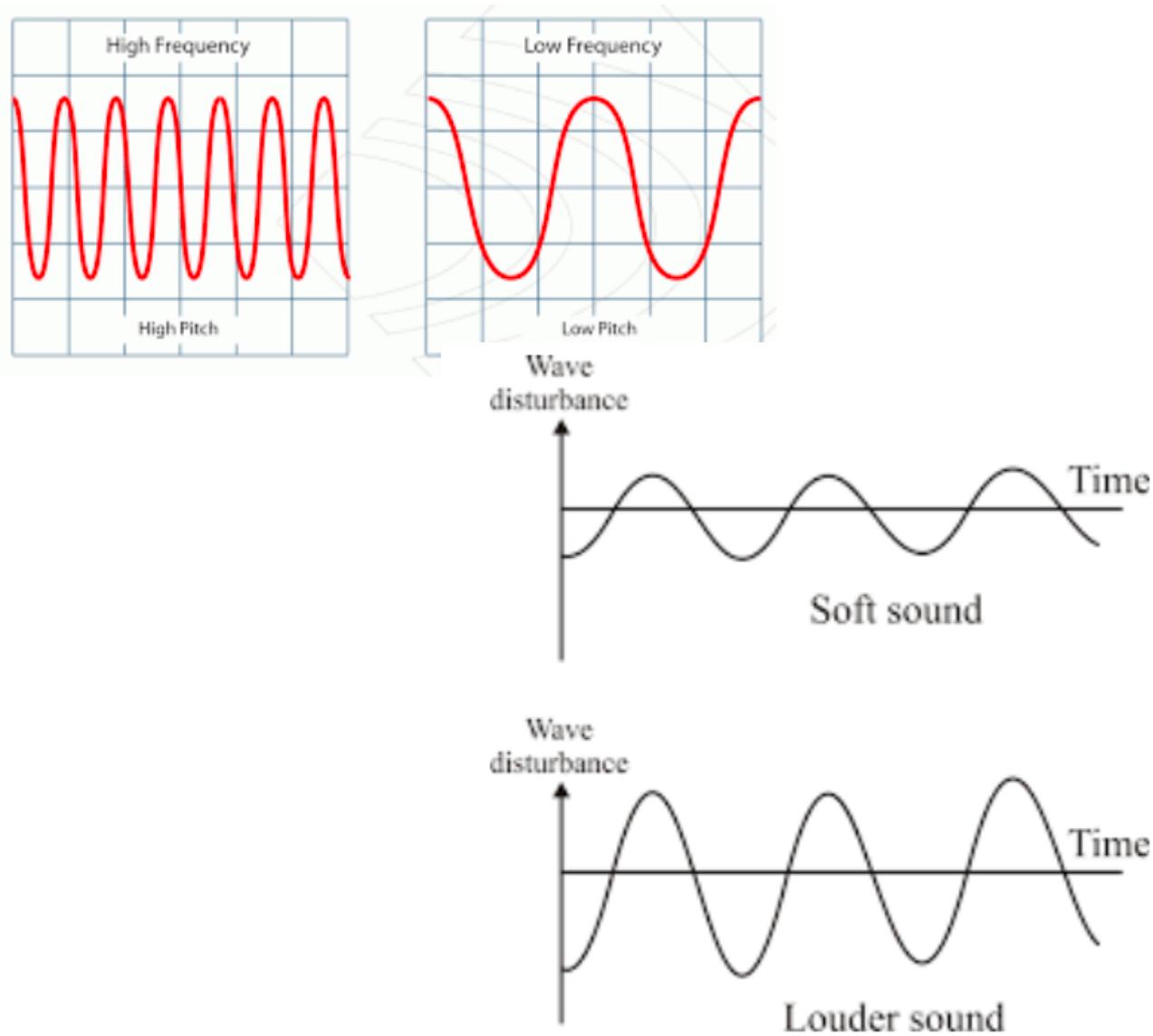
1. Using KISS page 17 and p 550-551 booklet complete the table in activity 8.6.4 on P552
2. Plan a long response “to evaluate the effectiveness of a hearing aid and a cochlear implant”



Hearing aid

cochlear implant

Sound waves:



Technology for Hearing Loss

Hearing Aids

Hearing aids are electrical devices that amplify sound in the environment for an individual to hear better by directing sound into the ear canal and are commonly used for people suffering from sensorineural hearing loss. It consists of a microphone, amplifier, and receiver. However, it does have some limitations such as the fact that it doesn't heal hearing-impaired individuals. And hearing aids for the canal cannot be used if the cochlear hair cell receptors are dead, as no amplification will help.

Bionic Ear

Bionic ears can directly stimulate the auditory nerve itself and bypass the activation of hair cell receptors. It requires surgical implantation, and consists of a microphone, speech processor, transmitter, receiver/stimulator, and electrodes. Limitations are that the surgery is costly from \$30k-50k, sound frequencies are also different to the environment so it will take a while to learn new sounds.

Bone Conduction Implants

Helps when the person's outer and middle ear structures are not functioning normally to detect sound waves. It consists of a microphone and a sound processor – transforms sound waves into vibrations which are then sent to the implant. Sound waves then travel to cochlea. Background noise received by external sound processor is lower than normal microphones, good as it removes unwanted noises. The limitation is that sometimes this might not be wanted in case of small sounds produced during dangerous events.

Technology for Sight Loss

Spectacles

Spectacles adjust refraction of light prior to striking the eye and undergoing refraction. Light strikes cornea and passes through to the lens of the individuals' eyes, resulting in light being refracted more towards retina. They are made from glass once but now plastic, light in weight and less of a chance of being broken. Contact lenses are also used to correct vision, are convex in shape and are fitted to match the curvature of the eyeball. These can be used for cosmetic purposes, sport, entertainment industry, and they can block UV radiation.

Refractive Laser Eye Surgery

Involves use of lasers to change curvature of cornea to alter refractive power to compensate for any visual defect in the eye. Lasers are computer operated.

Technology for Kidney Function Loss

Renal Dialysis

Renal dialysis involves the use of a semi-permeable dialysis tubing which is attached to an artery where the patient's blood is pumped into the tubing. Runs through dialysis fluid inside a machine, urea is not present in fluid. Salts are removed from blood during the process. Fluid flows in opposite of blood present in dialysing tubing, facilitates diffusion of urea out of the blood.

Kidney transplant

Usually used for end stage kidney disease, when a kidney is transplanted usually from a living donor.

Requires continue immunosuppressants