

# MODULE 5 - HEREDITY

**Reproduction** - refers to the production of offspring

## Mechanisms of Reproduction

Sexual	Asexual
<ul style="list-style-type: none"> <li>The fusion of genetic material from two parents which produce offspring that are different from their parents</li> </ul>	<ul style="list-style-type: none"> <li>Involves genetic material from only one parent and produces offspring that are genetically identical to their parent</li> </ul>

## Reproduction in Animals

	External Fertilisation	Internal Fertilisation
Advantages	<ul style="list-style-type: none"> <li>→ Lots of offspring can be produced</li> <li>→ Parents don't need to physically meet <ul style="list-style-type: none"> <li>◆ Therefore, the parents have longer survival rate</li> </ul> </li> <li>→ No need for parental care</li> </ul>	<ul style="list-style-type: none"> <li>→ Offspring have higher chance of survival</li> <li>→ Conserve resources</li> <li>→ They can choose their mate</li> </ul>
Disadvantages	<ul style="list-style-type: none"> <li>→ Chances of fertilisation are low because gametes are destroyed by environmental factors (eg. predators)</li> <li>→ Wasted resources and energy</li> <li>→ No guarantee any offspring will survive</li> </ul>	<ul style="list-style-type: none"> <li>→ Only a few offspring can be produced at once</li> <li>→ Parents must physically meet to reproduce</li> </ul>

**External** - generally occur in aquatic environments (eg. fish lay eggs in their environment)

**Internal** - Occurs in terrestrial environments and in mammals, reptiles and birds (eg. human mothers carry their offspring in their body)

EXTERNAL FERTILIZATION	INTERNAL FERTILIZATION
Male gametes swim to female gametes	Male gametes swim to female gametes
No copulation	Copulation occurs (e.g. sexual intercourse of male and female with transfer of gametes)
The male gametes are shed into a large space; there is less chance of fertilization	The male gametes are shed into a confined space; there is a greater chance of fertilization
Many female gametes are produced	Few female gametes are produced; this saves body materials
Zygotes develop outside male and female partners	Zygote can be retained inside the female's body for protection until it is fully developed
Most common in fish, amphibians and algae	Most common in land plants, reptiles, birds and mammals

## Reproduction in Plants

Asexual Reproduction	Sexual Reproduction
<ul style="list-style-type: none"> <li>→ Runners - long, thin, modified stems that grow on the surface of the soil</li> <li>→ suckers</li> <li>→ Vegetative propagation - where a branch from the parent plant is cut and replanted into the ground, where it will grow roots and establish itself as a new plant</li> </ul>	<ul style="list-style-type: none"> <li>→ POLLINATION - the transfer of pollen from one plant to another Birds, bees, insects, wind help with pollination</li> </ul>

## Reproduction in Fungi

Fungi reproduce according to their surroundings.

Sexual	Asexual
<ul style="list-style-type: none"> <li>• When conditions are poor, it reproduces sexually through spores that combine to form a new fungus. This increases genetic diversity, increasing chances of survival.</li> </ul>	<ul style="list-style-type: none"> <li>• When conditions are good, it reproduces asexually by budding or through spores.</li> </ul>

## Reproduction in Bacteria

Bacteria are prokaryotic, single celled organisms (eg. E.coli)

Bacteria reproduce through **binary fission**, where the cell replicates its DNA and the cells grow to about twice its original size. Resulting in the cytoplasm dividing and forming a new cell membrane.

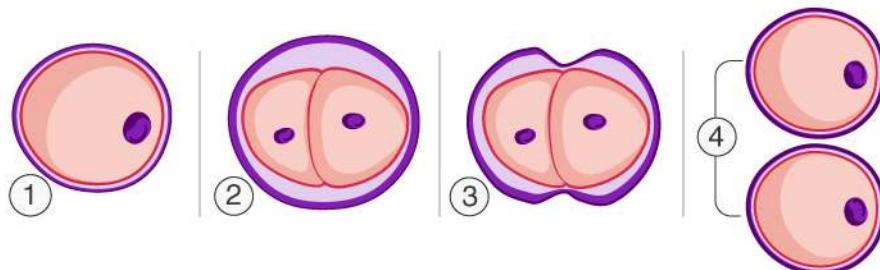
## Reproduction in Protists

Protists are eukaryotic, single celled organisms that do not classify as a plant, animal or bacteria.

Asexual reproduction - **binary fission**, sometimes budding

### BINARY FISSION

BYJU'S  
The Learning App



1 Parent cell

2 DNA Duplicates

3 Cytoplasm divides

4 Two daughter cells

# Advantages + Disadvantages of Reproduction Methods

## Sexual Reproduction

Sexual reproduction involves the fusion of genetic material from two parents. As a result, it produces offspring that are genetically unique. That is, offspring that are different from each other and from their parents.

Advantages	Disadvantages
<ul style="list-style-type: none"><li>Increases genetic diversity</li><li>The population is more likely to be able to adapt in response to change</li></ul>	<ul style="list-style-type: none"><li>Costs the parents time, energy and resources</li><li>Slower reproductive rate</li></ul>

## Asexual Reproduction

Asexual reproduction, on the other hand, involves a single parent, which produces a new individual from part of itself. Types of asexual reproduction include binary fission, budding, spores, cuttings and grafting.

It produces offspring that are genetically identical to each other and to the parent.

Advantages	Disadvantages
<ul style="list-style-type: none"><li>More efficient since the time and energy needed to produce offspring is much less compared to sexual reproduction</li><li>A population is able to reproduce quickly,</li></ul>	<ul style="list-style-type: none"><li>Low genetic diversity</li><li>Vulnerable to changing environmental factors, infections and pests</li><li>Population can easily decline</li></ul>

Feature	Sexual Reproduction	Asexual Reproduction
Number of Parents	Two	One
Genetics of Offspring	Unique	Identical
Type of Cell Division	Meiosis	Mitosis, binary fission, vegetative propagation and budding
Advantages	High genetic variation: the population is likely to be able to survive environmental change	High efficiency (less time and energy): population size can increase rapidly
Disadvantages	Low efficiency (costs time and energy): slow reproductive rate	Low genetic variation: the population is less likely to survive environmental change
Organisms	Animals, plants, fungi	Plants, fungi, bacteria, protists

especially in stable environments

- Evolution cannot occur

# Mammalian Reproduction

## Features of Fertilisation:

Fertilisation refers to the process by which the 2 gametes (egg and sperm) fuse together to form a single new cell that will develop into a new organism.

1. OVULATION: The female's body releases an egg from the ovary
2. The egg travels from the ovary to the fallopian tube (oviduct)
3. Male releases sperm during intercourse
4. FERTILISATION: The sperm fuses with an egg in the fallopian tube to form a **zygote**
5. Zygote forms strong outer membrane to stop more sperm from entering
6. Zygote will move to the uterus for IMPLANTATION

## Implantation:

Implantation refers to the attachment of the fertilised egg to the uterine lining. As the fertilised egg burrows deeper, it becomes enveloped by the lining of the uterus.

Uterus - where pregnancy takes place

Now that the zygote has developed into an embryo, it has more features including:

Amniotic sac - bag containing fluid which helps keep embryo at optimal temperature and provides cushioning

Placenta - provides nutrients and removes wastes

Umbilical cord - connects offspring to placenta

**Pregnancy** refers to the time the offspring spends within the female's body.

## Role of hormones:

Hormones are chemical messengers produced by the body, which travel in the blood to other cells where they have a specific effect.

Oestrogen

- Oestrogen is made by the placenta
- Stimulates ovulation
- Aids in blood flow to offspring
- Aids organ development
- Stimulates progesterone

Progesterone

- Progesterone is made by the ovaries and then by the placenta
- It stimulates the thickening of the endometrium
- Aids placenta function and relaxes the uterus
- Helps mother's immune system tolerate infant

Relaxin

- Loosens uterine muscles during pregnancy

Oxytocin

- Stimulates production of milk + Triggers uterine contractions

# Reproduction Manipulation in Agriculture

Agriculture is the growth of crops and animals for human needs.

## Selective Breeding:

Refers to the creation of organisms with certain desirable characteristics.

- Natural breeding - involves placing a male and female from the same species in an enclosed environment, and waiting for them to mate
- Artificial insemination - involves taking sperm from a male and inserting it directly into the female's reproductive tract
- Artificial pollination - involves taking pollen from one flower and inserting it directly into another flower using a small brush
- Cloning - involves creating a genetically identical copy of an organism using genetic technology techniques

## Positive Impacts:

- Increased sales due to increased population or improved food quality
- Increases resistance to certain pests and disease
  - Farmers spend less money on pesticides and medicines and gets more produce in return
  - Less environmental contamination

## Negative Impacts:

- Reduces biodiversity
  - As 'undesirable' traits are bred out of the gene pool, variation in a species decreases
  - Population is more likely to suffer from changes in the environment

Atom Thesis: The impact of manipulating reproduction in agriculture is huge and positive, so long as the threat of reduced biodiversity is managed.

# DNA + RNA Structure

DNA - deoxyribonucleic acid

RNA - ribonucleic acid

## DNA:

DNA is a type of nucleic acid that is responsible for storing genetic information in cells.

Acts as an 'instruction manual' since it contains information needed for growth, survival and replication.

Eukaryotes:

- Relatively large amount of DNA
- DNA is bound (wrapped around by histones)
- Forms linear chromosomes
- Chromosomes reside in the nucleus

Prokaryotes:

- Relatively small amount of DNA
- DNA is unbound
- Have a single, circular chromosome
- Chromosome resides in the cytoplasm

## DNA STRUCTURE:

Consists of building blocks called (deoxyribonucleotides) nucleotides, which include a nitrogenous base, a deoxyribose sugar and a phosphate group. Deoxyribonucleotides join together, forming a long chain. The two chains twist around each other to form a double helix structure.

Summarised: Polynucleotide strands are held together by hydrogen bonds between complementary bases.

Two polynucleotide strands twist to form a double helix structure.

Nitrogenous bases are connected by hydrogen bonds. Each base has a complementary base pairing:

- Adenine = Thymine
- Guanine = Cytosine

This ensures the structures are the same length and the proportions of the bases are maintained.

## RNA:

A type of nucleic acid that is responsible for interpreting genetic information from DNA into proteins.

## RNA STRUCTURE:

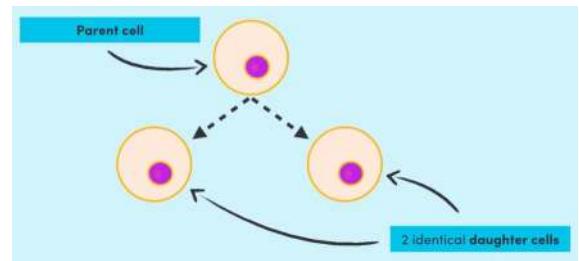
- Consists of nucleotides, which contain a sugar, phosphate and nitrogenous base.
- RNA is single stranded
- There is a specific type of nucleotide in RNA called ribonucleotide
- DNA and RNA have most of the same bases, but RNA never has THYMINE. Instead it has URACIL

Nitrogen bases in RNA include:

- Adenine = Uracil
- Guanine = Cytosine

# Mitosis

**Definition:** Mitosis is a type of cell division where **1 parent cell** divides **ONCE** to produce **2 IDENTICAL** daughter cells.



## Process:

Within the nucleus, DNA is wrapped with proteins which is referred to as **Chromatin**.

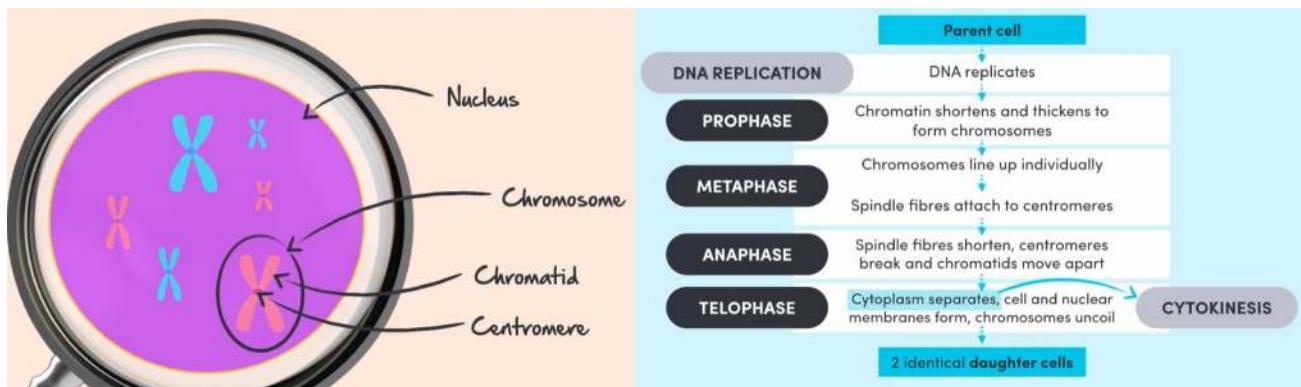
The first step of mitosis involves the replication of DNA (chromatin).

The two identical chromatin pieces are still joined at the middle, and they quickly shorten and thicken.

When chromatin is all coiled up into the X-Shape, it is now referred to as a **chromosome**.

Each chromosome has two strands, called chromatids which are the replicated chromatin pieces connected together at the centre by a **centromere**.

Inside a normal body cell, there are **two sets of chromosomes which code for the same genes** (one from the mother and the other from the father). These alternative forms of the same genes are called **alleles**.

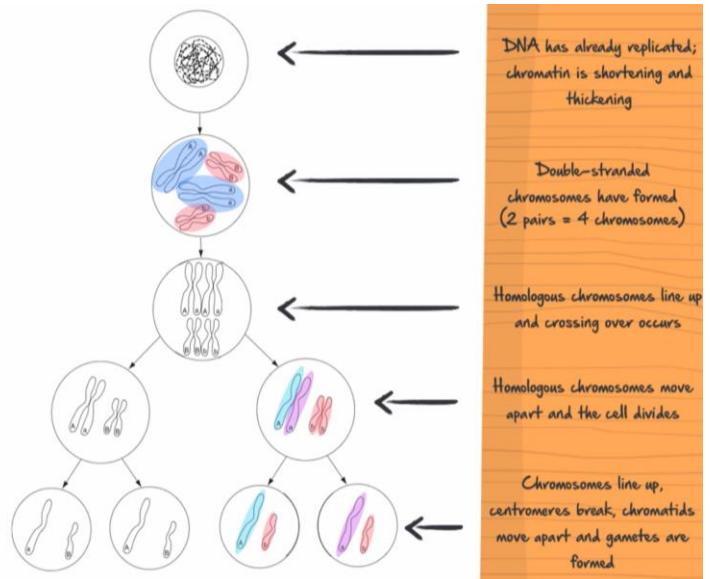


## (IPMATIC) I-Interphase (preparation phase)

## Why is Mitosis Important?

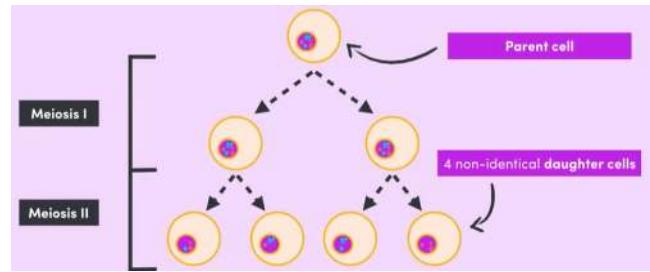
Mitosis is important because it creates new body cells that are needed for growth, repair and maintenance.

- Important for the **individual** - It allows the healing of injuries, growth and survival
- Important for the **species** - increases an organism's chances of reproducing



# Meiosis

**Definition:** Meiosis is a type of cell division where **1 parent cell** divides **TWICE** to produce **4 NON-IDENTICAL** daughter cells.



## Process:

Similar to mitosis but cell division occurs twice (IPMATIC happens twice). Within meiosis two significant things happen, random segregation and crossing over.

**Random Segregation** - the chromosomes line up with their matching pairs in a random order

**Crossing Over** - the exchange of genes between homologous chromosomes

## Why is Meiosis Important?

Meiosis allows for the **continuity of a species** because it means that organisms can produce offspring with new gene combinations.

This allows chances of **Evolution** (natural selection) since a population with new gene variations have a better chance of survival during environmental changes.

FEATURE	MITOSIS	MEIOSIS
DIVISIONS	1	2
DAUGHTERS	2	4
CHROMOSOMES	2n (diploid) 6 chromosomes	n (haploid) 3 chromosomes
GENETICS	Identical to parent cell and other daughter cell	Not identical to parent cell or other daughter cells
CROSSING OVER	No	Yes
ROLE	Growth, repair, maintenance	Continuity of species (reproduction)

The diagram models the process of meiosis.

Explain the structure and behaviour of chromosomes in the first division of meiosis.

Include detailed reference to the model. (5 marks)

**1. Crossing over**      State cause      State effect

during the first division of meiosis, homologous chromosomes pair up and undergo crossing over, which is where sections of DNA are exchanged to create genetic variation through new allele combinations. This is represented by the mixed grey and white chromosomes in the diagram after the first division.

Link to model

To show we are addressing the whole question!

Link to genetic variation

**2. Random segregation**      State cause      State effect

The homologous pairs then line up on the equator and are randomly separated into the two daughter cells (random segregation). Each cell contains half the original number of chromosomes, with one chromosome from each homologous pair, as seen in the diagram. This creates variation through the unique combination of chromosomes in each cell.

Link to genetic variation

Link to model



# Sources of Genetic Variation in Sexual Reproduction

**Definition:** Genetic variation is a term used to describe the differences between the genomes of individuals of the same species

Basically, it refers to the differences in the DNA of a group of similar organisms. Similar enough to breed although they do not have the exact same genes. There are usually lots of different versions of the same genes (alleles) in the gene pool.

## **Recap Meiosis - Random Segregation + Crossing Over:**

The homologous (matching) chromosomes have paired up along the equator randomly.

Independent Assortment - Pairs of homologous chromosomes arrange independently to each other. As a result, it is referred to as random segregation where the chromatids segregate causing the set of chromosomes in the daughter cells to be random.

Due to the different variations of chromosomes, it results in greater genetic variation

Crossing over is when 2 homologous chromosomes swap sections to produce new gene combinations. It is a critical source of genetic variation because it changed the genetic composition of the chromosomes.

## **Fertilisation:**

During fertilisation, a paternal gamete and a maternal gamete fuse to form the first cell of the new organism. Fertilisation contributes to genetic variation because it allows for different gametes (containing different alleles) to combine.

There are lots of unique organisms in a species, which all have a variety of unique gametes (due to meiosis). Any of these gametes can combine during fertilisation to form different zygotes.

## **Mutations:**

A mutation refers to a change in the base sequence of an organism's DNA.

A change in the DNA sequence results in the formation of different genes which ultimately causes different genotypes and phenotypes.

Mutations are the ultimate source of new alleles in a species.

# Genotype + Phenotype

## Genotype:

'Genotype' refers to an organism's genetic makeup for a particular characteristic. That is, it's the set of alleles in an organism's DNA which code for a trait.

Typically, an individual's genotype for a trait consists of two alleles: one inherited from their mother, and one from their father.

In humans, genotype is determined at fertilisation, when a sperm and egg fuse to form the first cell of a person, which we refer to as a zygote.

## Phenotype:

'Phenotype' refers to an organism's physical expression of a particular characteristic. That is, the observable or expressed characteristics of an organism.

An organism's phenotype for a particular characteristic is determined by two things: which alleles are present (the genotype) as well as environmental factors.

The relative influence of genotype and environmental factors varies depending on the characteristic. So, the phenotype of some traits is entirely controlled by the genotype, whereas the phenotype of other traits is affected by genes and certain environmental factors.

*Most offspring resemble their parents in a number of characteristics, but there are often some characteristics in the offspring that are unexpected.*

Explain, using examples, how genetics and environment can affect the phenotype of individuals. (8 marks)

Definition

Genetics

Split answer into subheadings

A gene is a section of DNA whose base sequence codes for a specific protein.

1. Sex linked inheritance

Genetics affect phenotype in sex-linked inheritance. Since the Y-chromosome carries fewer genes than the X-chromosome, males only require a single X-linked recessive gene to inherit the sex-linked phenotype. Example: red-green colour blindness. A boy could be born with red-green colour blindness even though neither parent may have the condition.

Provide numbered dot points

2. Mutations during meiosis

Genetic mutations can alter the genotype to produce unexpected offspring without affecting the parents. Example: down syndrome. This is caused when homologous chromosomes are incorrectly separated during meiosis, changing physical characteristics of the offspring despite neither parent having the condition.

*Most offspring resemble their parents in a number of characteristics, but there are often some characteristics in the offspring that are unexpected.*

Explain, using examples, how genetics and environment can affect the phenotype of individuals. (8 marks)

Definition

Genetics

1. Sex linked inheritance

2. Mutations during meiosis

Split answer into subheadings

The environment can generate unexpected offspring phenotypes through mutation or resource availability.

1. Mutations due to mutagens

Environmental mutagens can alter DNA sequences and cause phenotypic changes not expressed by the parents. Example: UV radiation. This can mutate regulatory DNA sequences, causing uncontrollable cell growth expressed as skin tumours.

This shows we have

Link to the question

answered the whole question!

2. Resource availability

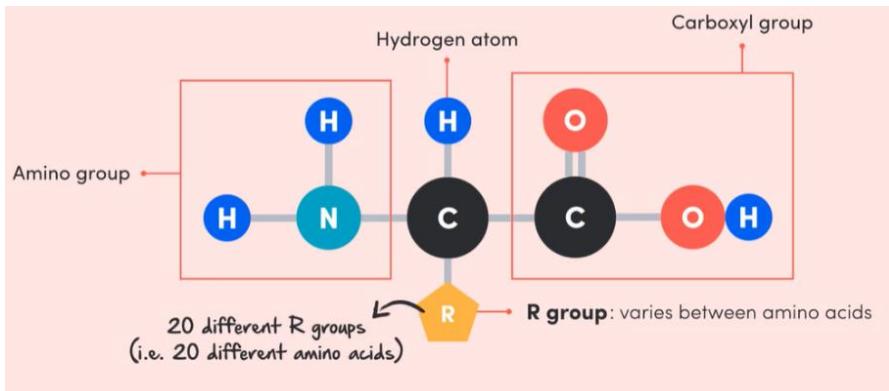
As parents and offspring have access to different resources, the offspring's phenotype can be expressed differently. Example: Himalayan rabbits. The rabbits are homozygous for a heat sensitive enzyme which is only activated at low temperatures. This activation causes black fur to grow, generating a different phenotype to the parent's white fur.

# Proteins

## Structure:

A protein consists of a chain of building blocks called amino acids, which are linked together via peptide bonds. A long chain of amino acids is called a polypeptide chain.

Basic structure of an amino acid:

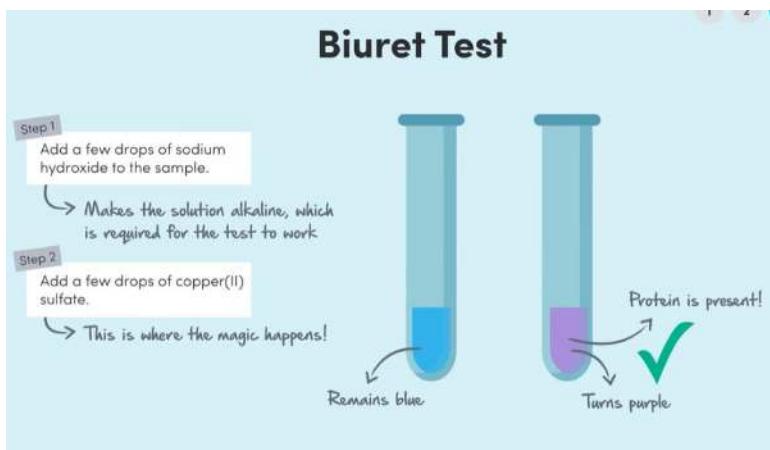


## Function:

Proteins are a major component of every cell. All the proteins within a cell are referred to as a PROTEOME.

Proteomes are split into four categories:

- 1) Structural proteins - maintain cell shape and make our connective tissues → important for growth, repair and maintenance of tissue (Eg. collagen)
- 2) Enzymes - biological catalysts. They act on specific substrates to either break it down or to combine it with another substrate to make it more complex
- 3) Hormones - Proteins which are secreted into the blood by endocrine cells (glands) → travel to target tissues, where they cause a change in activity
- 4) Immunity - Antibodies are the proteins involved in the immune response → Antibodies react with antigens (foreign particles) to help remove them from the body.



# Protein Synthesis

## Introduction to Genes:

The order of bases in DNA acts like an ‘instruction manual’ for when the cell wants to make proteins. DNA is split up into genes. Genes refer to a portion of DNA that tells the cell how to make a specific protein. Genes determine the order and type of amino acids.

Codons are a set of three bases which code for one particular amino acid in the polypeptide chain. They are very specific as it only codes for a specific amino acid. Codons do not overlap.

The order of codons and the bases within them determine which amino acids are produced and in what order. There are special codons that say when to start and when to stop reading the code (Start codon + Stop codon).

## Transcription:

Basically, this is where an mRNA copy of a gene is made when DNA is used as a template.

This takes place in the nucleus.

### INITIATION

1. RNA polymerase attaches to DNA at the desired gene and separates the strands to expose the nucleotides in that region. This breaks the hydrogen bonds between the nucleotide bases, causing the double helix to separate.

Only the section with the gene is pulled apart, not the whole DNA strand

### ELONGATION

2. One strand is used as a template to make an mRNA strand identical to the other strand through complementary base pairings. (Eg. original is AGCT, mRNA will be UCGA)
3. Free floating nucleotides pair with their complementary bases on the template strand.  
RNA polymerase adds RNA nucleotides one at a time as the DNA strands rejoin behind it. Hydrogen bonds are being formed between RNA and DNA bases as the bases match up.

### TERMINATION

4. The RNA polymerase reaches a stop codon and lets go of the DNA strand.
5. In **eukaryotes**, introns are spliced out of the strand and exons are stuck together to form the final mRNA strand. Exons remain because they are expressed. Introns do not code for proteins.
6. mRNA molecules leave the nucleus and enter the cytoplasm through a nuclear pore

## Translation:

In this stage, nucleic acid is translated into a protein. A polypeptide chain is formed according to the sequence of codons.

### INITIATION

1. The mRNA from the nucleus travels through the cytoplasm and attaches to a ribosome at a particular start codon  
**Ribosome:** An organelle found in the cell’s cytoplasm, or attached to the endoplasmic reticulum in eukaryotes
2. Free-floating tRNA molecules found in the cytoplasm then come into the scene. Each tRNA molecule has a specific anticodon sequence  
**Anticodon:** A triplet of bases that corresponds to a specific mRNA codon

## ELONGATION

3. The tRNA molecule with the correct anticodon pairs up with the mRNA in the ribosome  
Each tRNA molecule is bound to a specific amino acid. Specific codon → specific amino acid
4. A second tRNA molecule attaches to the next codon on the mRNA strand
5. The ribosome catalyses the formation of a peptide bond between the two amino acids  
The first tRNA molecule moves away from the ribosome, leaving its amino acid
6. The ribosome continues to move along the mRNA until it reaches a stop codon  
By this point, it has formed a polypeptide chain but is still bound to the ribosome by a single tRNA molecule

## TERMINATION

7. The polypeptide chain and mRNA strand are released from the ribosome through a signal in the stop codon, telling the tRNA strand to let go

### From polypeptide to protein:

Since a protein is formed by one or more polypeptides, the polypeptide chains need to be coiled and folded into functional protein structures.

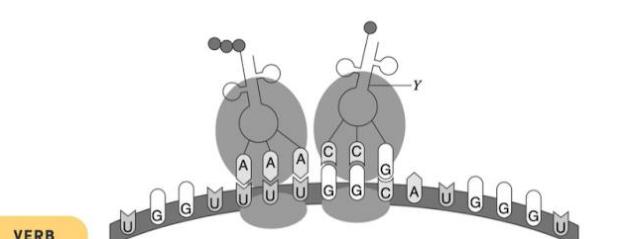
Protein structures include primary, secondary, tertiary and quaternary structures:

- Primary : a linear sequence of amino acid in the polypeptide chain, it provides information on how proteins will fold
- Secondary : the folding and coiling of the peptide chain in order to form bonds between different chains later. A common structure is the alpha helices, beta sheets and random coils
- Tertiary : a combination of alpha helices and beta sheets by hydrogen bonds  
In extreme conditions, the protein structure will lose its shape and denature, losing its function.
- Quaternary : formed when more polypeptide chains join to create a single functional protein.  
(e.g. hemoglobin)

### Importance of mRNA and tRNA in protein synthesis:

- Since DNA is stored within the nucleus, mRNA is essential in transporting the genetic information out of the nucleus to the ribosomes in order to make proteins.
- mRNA contain start and stop codons that initiate and terminate the synthesis of polypeptide chains
- tRNA ensures that specific amino acids are connected to form the correct polypeptide chain. It ensures the correct structure and function of proteins.

This diagram shows part of the process of protein synthesis.



Outline how the structure Y enables information from DNA to be translated into a specific polypeptide. (4 marks)

Look over the stimulus first!

Identify structure in stimulus

Split answer into numbered dot points

Start with DNA & transcription to provide context

Y is a tRNA molecule. The process of DNA translation into a polypeptide is as follows:

1. The sequence of nitrogenous bases in DNA codes for amino acids through three-base codons.
2. mRNA transcribes code by binding to complementary bases and exits the nucleus, entering the cytoplasm.
3. At ribosomes, tRNA (with attached amino acids) translates code by binding to mRNA codons via complementary anticodons (e.g. AAA-UUU and CCG-GGC in diagram).
4. A ribosome holds tRNA molecules in order specified by codons whilst peptide bonds form between adjacent amino acids to form a polypeptide.

As such, the order of tRNA molecules determines the order of amino acids and the type of polypeptide produced.

Link back to question



# Inheritance

Inheritance explains how characteristics are passed from one generation to the next, based on the transfer of DNA.

## Alleles:

Alleles are different versions of the same gene, and are found at the same place on homologous chromosomes.

- Dominant allele is always expressed in the phenotype
- Recessive allele is only expressed in the phenotype when there is no dominant allele

If an organism has 2 different alleles for a particular gene, it is referred to as HETEROZYGOUS

If they have 2 identical alleles for a particular gene, it is HOMOZYGOUS

## Punnett Squares:

Punnett squares are tables which are used to predict the possible genotype and phenotype of the offspring produced by a particular male and female.

Key term	Definition
Gamete	Gametes are sex cells (in animals: sperm and ovum; in plants pollen nucleus and ovum).
Chromosome	Chromosomes are thread-like structures of DNA, carrying genetic information in the form of genes. They are located in the nucleus of cells.
Gene	Genes are short lengths of DNA found on chromosomes. They code for specific proteins.
Allele	Alleles are different versions of a particular gene.
Dominant	A dominant allele is always expressed, even if only one copy is present.
Recessive	A recessive allele is only expressed if two copies are present (therefore no dominant allele present).
Homozygous	If the two alleles of a gene are the same, we describe the individual as being homozygous (homo = same).
Heterozygous	If the two alleles of a gene are different, we describe the individual as being heterozygous (hetero = different).
Genotype	The combination of alleles that control each characteristic is called the genotype.
Phenotype	The observable characteristics of an organism (seen just by looking—like eye colour, or found through testing—like blood type) is called the phenotype.

# Autosomal and Sex-linked Inheritance

Humans have 23 homologous pairs of chromosomes (46 chromosomes in total) and each pair codes for the same genes.

- 23 from mother
- 23 from father

## Autosomes:

- Humans have 22 pairs of autosomes
- Eg. eye colour and blood type

## Sex chromosomes:

- Determine the sex of an organism:
  - Female - 2 similar chromosomes (XX)
  - Male - 2 dissimilar chromosomes (XY)
- Still codes for some characteristics that are not sex-specific
- Eg. breast development in women and Adam's apple development in men

## Autosomal Inheritance:

Patterns in the expression of characteristics which are found in autosomes.

- Dominant/ Recessive
  - Dominant allele is always expressed
    - Homozygous dominant
    - Heterozygous
  - Recessive allele is only expressed only when there is no dominant allele
    - Homozygous recessive
- Co-dominant
- Incomplete

## Sex-linked Inheritance:

Patterns in the expression of characteristics which are found on sex chromosomes (X&Y).

Sex-linked inheritance is different to autosomal inheritance due to two facts:

1. The Y chromosome has fewer genes than the X chromosome
2. Males only have one copy of the X chromosome
  - If a man inherits a faulty allele on this X chromosome, it will have to be expressed

X- Linked	Y- Linked
<p>Transmission of genes which are found on the X chromosome.</p> <p><b>Recessive:</b></p> <ul style="list-style-type: none"><li>• Affect men more than women</li><li>• Higher chance of a man inheriting one faulty X chromosome</li></ul> <p>→ Eg. red/green colorblindness</p> <p><b>Dominant:</b></p>	<p>Transmission of genes which are found on the Y chromosome.</p> <ul style="list-style-type: none"><li>• Only passed from father to son</li><li>• Never seen in females</li></ul>

- Affect genders equally

## Co-dominance and Incomplete Dominance

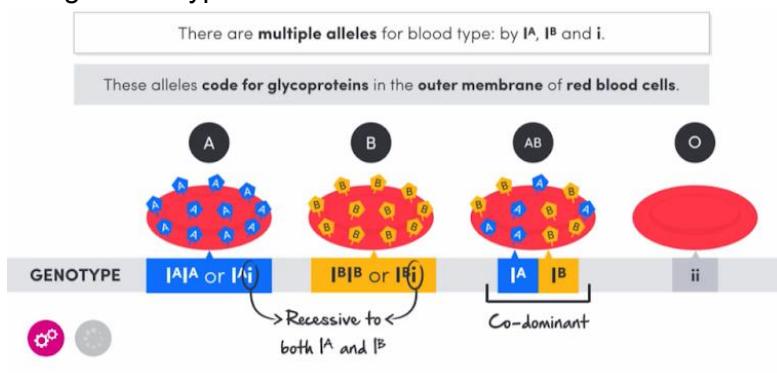
**Co-dominance:** refers to a situation in which both alleles are expressed in the phenotype

- ★ During exams, NEVER use the words “mixing” or “blending” since the DNA has not mixed to create a new allele
- ★ Common exams examples - Roan cattle, human blood types, chickens, tabby cats

→ Eg. Roan cattle - shows both red and white fur



→ Eg. Blood types



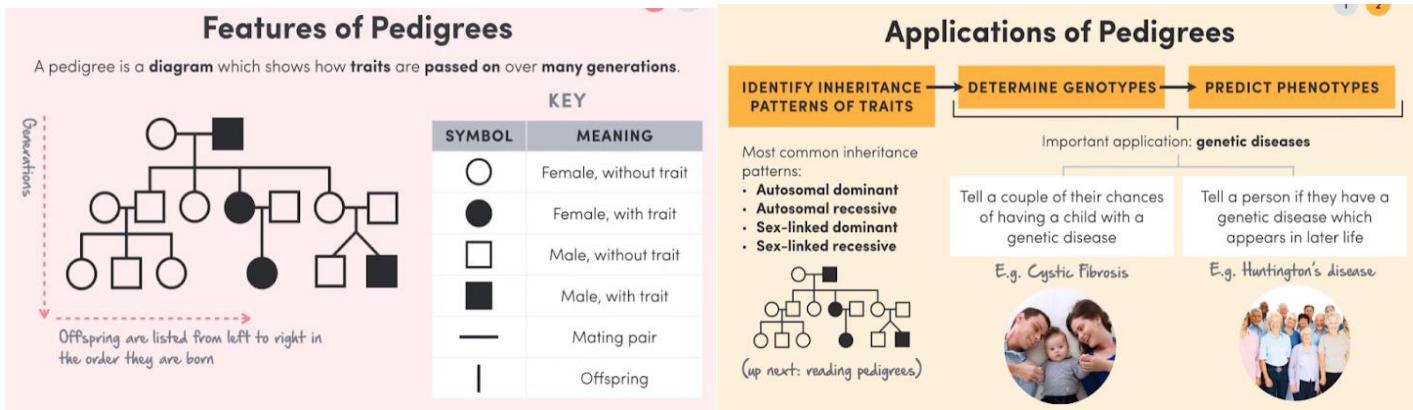
**Incomplete Dominance:** The dominant allele is only partially expressed

- It doesn't completely control the phenotype of a heterozygous organism
- The heterozygous phenotype is ‘in between’ the homozygous phenotypes

→ Eg. Snapdragon flowers



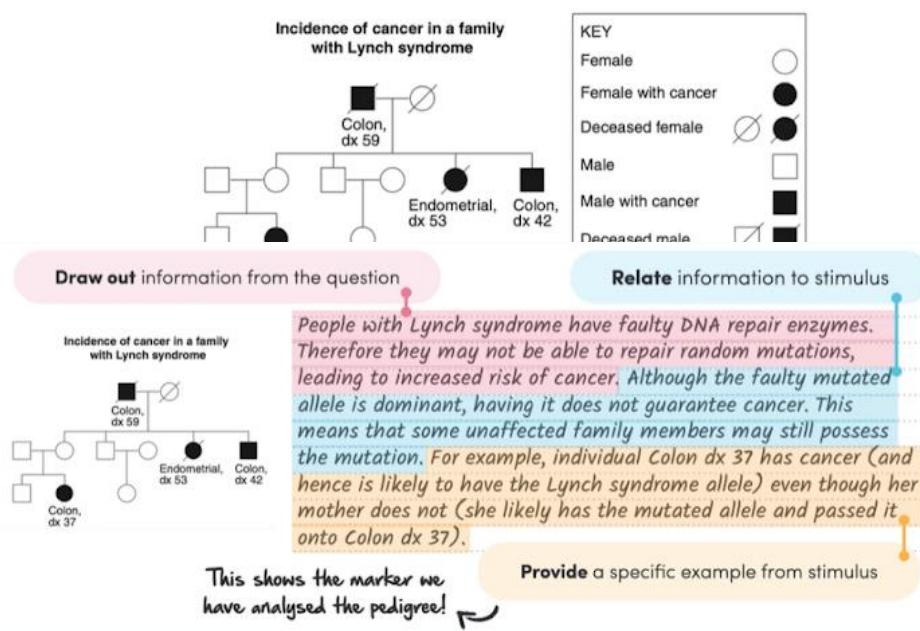
# Pedigrees



## Reading Pedigrees:

- Check the key and identify the affected individuals
- Looking at the sex to determine whether the trait is autosomal or sex-linked
  - Autosomal - no. of affected females is around the same no. of affected males
  - Sex-Linked - almost all affected individuals are males
- Look at the generations to determine whether it is dominant or recessive
  - Dominant - usually appears in every generation (because every affected person must have an affected parent)
  - Recessive - if the parents are affected, all children have the affected phenotype
- Confirm the estimate - double check by working backwards and applying the theory using (eg. AA, aa, Aa)

Lynch syndrome is an autosomal dominant condition that is the result of a mutation of a DNA repair gene. This mutation interferes with the ability of DNA to repair itself. The incidence of cancer in a family with Lynch syndrome is shown in the pedigree.



# Allele Frequency

**Allele frequency** is the relative proportion of a particular allele in a population.

So, it can be represented using the following equation:

$$\text{Allele frequency} = \frac{\text{Number of an allele in a population}}{\text{Total number of alleles in the population}}$$

- Allele frequency refers to the relative proportion of a particular allele in a population.
- You can work out allele frequencies by taking a sample of the population, as long as that sample is big enough for us to say that it is likely to be representative.
- Allele frequencies can be calculated from information about alleles, or by looking at phenotypes so long as we know the relationship between genotype and phenotype.

## Factors that affect allele frequency:

- Isolation
- Migration
- Natural selection
- Artificial selection
- Genetic drift is the natural change of allele frequency, which comes from random events such as natural disasters - this has major impacts on smaller populations. Larger populations receive less impact

## Predicting Speciation:

Typically, speciation occurs when there is a barrier to gene flow. There are lots of different kinds of barriers, but an example would be a valley created by an earthquake which splits a population in half. If populations stop interbreeding with each other, then their DNA will become less and less similar. We can say that the gene flow between species has stopped. When there is little or no interbreeding between populations, new mutations which arise in one population will not spread to the other. Genetic variants might be lost from one population, but not the other. Overall, this means that allele frequencies will change.

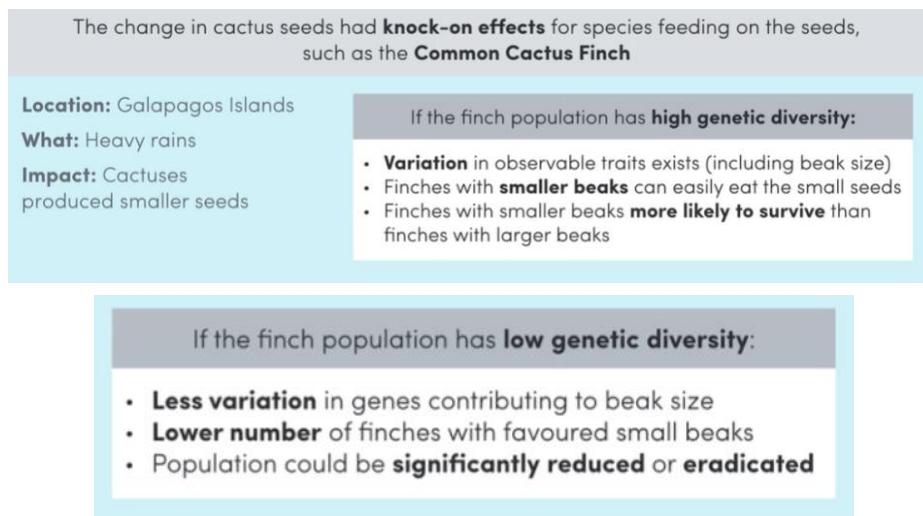
- There are several evolutionary processes which can cause populations to move away:
  - Natural selection
  - Preferential mating
  - Migration
  - Mutations
  - Random events
- If the allele frequencies in two populations are different, there is probably a barrier to gene flow.
- If we measure frequency data for enough alleles, we may be able to work out if two populations have stopped interbreeding, making them different species

# Population Genetics in Conservation

## Measuring Extinction Risk:

Populations with greater genetic diversity withstand environmental changes better.

Conservation scientists measure population genetics to calculate the risk of extinction and to better understand the ecosystem.



## Monitoring Inbreeding:

Inbreeding refers to when closely related individuals breed together.

Closer related individuals have similar ancestors, so are more likely to inherit the same unhealthy alleles.

Unhealthy alleles are often recessive → inheriting two copies of the unhealthy allele led offspring to die at younger ages → high mortality rate among the inbred population → decline in population

This occurs when individuals only have access to a small range of potential mates.

→ May occur if population size suddenly falls (eg. due to environmental catastrophes)

→ May occur if population is split into smaller groups (eg. due to habitat fragmentation)

Comparing the genomes and a random population sample allow us to monitor inbreeding by:

→ allowing us to determine how closely related the population members are

→ allowing us to measure the allele frequencies for unhealthy mutations

## Investigating Speciation:

Speciation refers to the formation of new species.

Species is defined as a group of organisms that interbreed to produce fertile offspring.

Cryptic species: groups of originally thought to be same species, but are actually different

During speciation, populations stop interbreeding, preventing DNA from being shared between populations, creating a barrier to gene flow.

→ Alleles may be lost in one population and not the other

→ New alleles may arise in one population and not the other

Population genetics allows us to compare the allele frequencies within and between populations → if allele frequencies are becoming very different, it suggests speciation is occurring

## Population Genetics in Disease Inheritance

### Diseases affected by multiple genes:

- Many genetic disorders are affected by more than one gene.
- Polygenic inheritance refers to when the inheritance of an observable trait is determined by many genes.
- Genes showing polygenic inheritance often have environmental influences
- Identifying the regions of DNA associated with a genetic disorder shows:
  - Who is likely to be affected?
  - What goes wrong in the cell to cause the disease?
  - How can we treat the disease?

### Genome-Wide Association Studies (GWAS):

- GWAS involves taking the DNA of an individual, and looking for certain variations
- Rather than looking through the entire genome, GWAS involves a targeted search for bases that are known to be highly variable (**Single nucleotide polymorphisms** AKA SNP) in the population
  - **SNPs**- by comparing the SNPs of people with a disease to those without, we can identify which SNPs are associated with a disorder
  - SNPs are more common than mutations causing rare genetic diseases

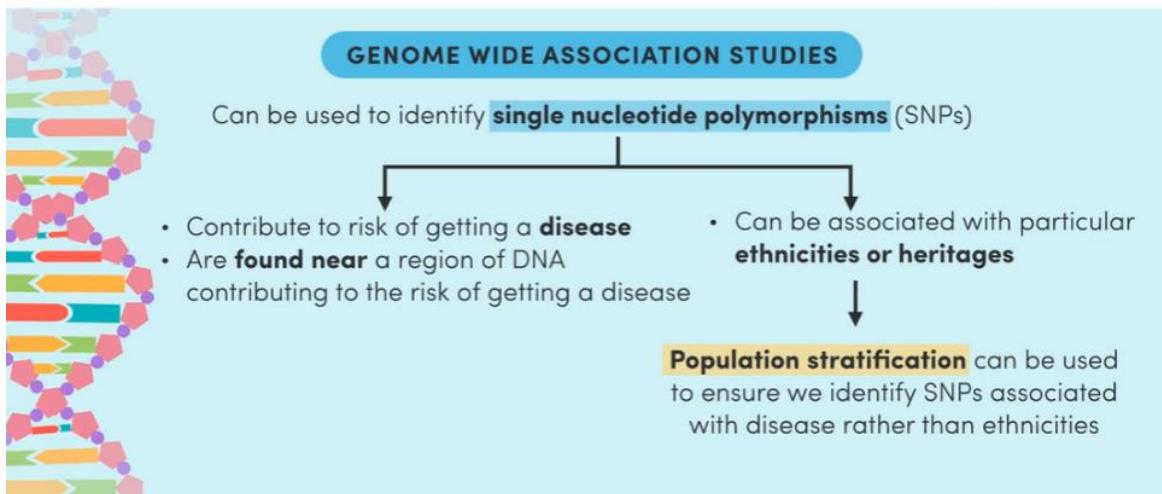
### Limitations of GWAS:

- Population stratification refers to when disorders are more common in people with a particular ancestry or ethnicity
- People of shared ethnicity are more likely to share certain SNPs
- However, these shared SNPs may not be associated with having a disease
  - May just be associated with ancestries

### How do we avoid confusing SNPs associated with heritages and SNPs associated with disorders?

When carrying out a GWAS, we should not just take random samples from the affected (with disease) and the unaffected populations to compare. When sampling, it is essential to control for ancestry.

Many diseases show **polygenic inheritance**, meaning their inheritance is shown by **many genes**



## Population Genetics in Human Evolution

### Primate Evolutionary Relationships:

- Humans are scientifically classified as Homo Sapiens
- We can base our ideas about the evolutionary relationships of different species on their observable characteristics as well as DNA evidence
- Analysing DNA can inform evolutionary relationships

### DNA and Evolutionary Relationships:

- The more DNA is collected and analysed, the more accurate the understanding of primate evolution
- On average, each base pair of primate DNA has one in a billion chance of mutating per year
- If we know the rate of DNA mutation, we can work out when the last common ancestor of two species or populations lived. This allows us to work out the migration of species.

### Studying Human Migration:

- We can work out how long-ago different groups of humans migrated using population genetics
- To do this, we look at how genetic markers vary across people in different populations using databases such as the Human Genome Project
- The amount of difference between the genetics of two people increases with the distance apart they live
- Identifying the rate of DNA mutation means we can work out the rate that new genetic markers arise, and how long-ago populations last shared a common ancestor

# Polymerase Chain Reaction (PCR)

## What is it?

- PCR is a technique used to amplify DNA, in vitro
  - PCR involves making lots of copies of a specific region of DNA  
(Usually necessary if we want to test, analyse or use a DNA sample)
  - PCR is performed in a test tube, rather than a living organism

## PCR Materials:

- Template
  - DNA sample which is going to be copied in PCR (can be sourced from a crime scene, or a person being tested for a genetic disease)
- Free Nucleotides
  - Building blocks of DNA
- Heat-Stable DNA Polymerase
  - Enzyme which catalyses DNA synthesis, according to the base sequence of the template  
Since PCR involves large temperature changes, it's important that we have a DNA polymerase that can survive the changes - 'heat-stable'  
→ otherwise, a new DNA polymerase enzyme would be needed after each cycle
- Primers
  - Short, single stranded pieces of chemically synthesised DNA which flank the target region
- Buffer
  - Liquid into which all of the other components are added; prevents sudden pH changes

## Steps in PCR:

To do a PCR, the mixture is placed in a thermal cycler, so that the mixture can be heated and cooled in a controlled way.

PCR uses variations in temperature to control the replication process in three steps:

1. Denaturation (denature)
  - PCR reaction mixture is heated to 95 degrees Celsius
  - DNA is denatured, i.e. separates to form two single strands which act as templates for the next step
2. Annealing
  - PCR reaction mixture is cooled to 55 degrees Celsius
  - Allows primers to anneal (= bind) to the template DNA
3. Extension
  - PCR reaction mixture is heated to 72 degrees Celsius

- DNA polymerase moves down the template, synthesising new DNA

## Why is it important?

Scientists often need large amounts of DNA if they want to use it in DNA sequencing, DNA profiling and/ or genetic testing

Outline how the polymerase chain reaction can be used to amplify DNA sequences. (3 marks)

List the steps of the process

This gives the marker a logical structure to follow!

The three steps of PCR are: denaturation, annealing and extension.

1. **Denaturation** DNA is heated and the complementary strands are separated. After this, the mixture is cooled.

2. **Annealing** primers bind to sections of each separate DNA strand.

3. **Extension** polymerase catalyses the complementary bonding of free nucleotides along the template DNA strands.

The process is then repeated multiple times to reproduce (amplify) DNA.

→ This shows the marker we know what we're talking about!

[Link back to the question](#)

[Sketch each step in general terms](#)

# Gel Electrophoresis

Gel electrophoresis is used to separate and visualize DNA fragments according to their size. Basically, it is just a fancy term for a filtering system.

## Steps in Gel Electrophoresis:

- ★ Remember! Phosphates in DNA have a negative charge → DNA has an overall negative charge → crucial for gel electrophoresis

### 1. Prepare the set up

- Pour liquid agarose gel into a mold
- Insert a well comb
  - It creates identical small holes in the agar
- Once the mold has set, place it into a gel box
  - The gel box has a positive electrode and a negative on the other end.
  - The wells are made at the negative end of the gel box.
- Immerse gel with buffer (can conduct electricity)

### 2. Load DNA samples and DNA ladder

- Each DNA sample is transferred into its own well
  - DNA can be sourced from crime scenes or patients
  - DNA samples are usually amplified first (using PCR) before it is tested in a gel, this is so there are more things to see
- A DNA ladder is added into one well
  - DNA ladder is a mixture of known DNA fragments, each of which has a specific size
  - Allows the determination of the size of the DNA fragments (by comparing to the ladder)

### 3. Run the gel

- Turn on the power
  - a current runs through the gel
- DNA migrates through the gel, towards the positive pole

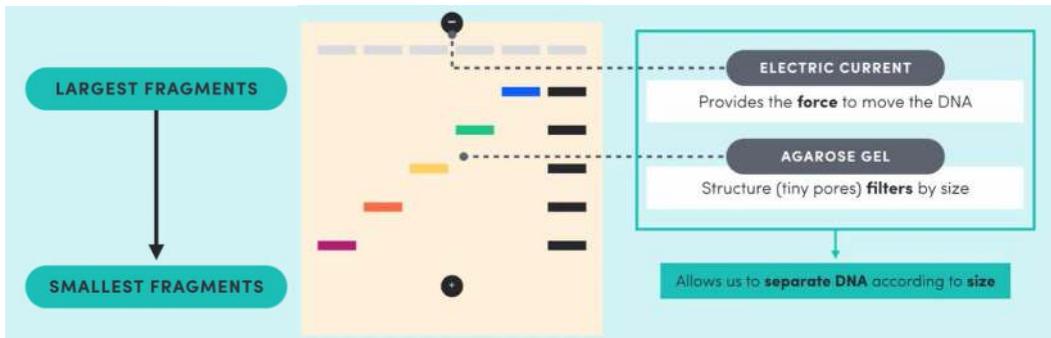
### 4. Visualising the DNA fragments

- Add a dye to stain the DNA fragments
  - Dye may be added to liquid agarose, before it is poured into the mould or to the gel, after it has been run
  - this allows us to see the DNA fragments

If all of the DNA is negatively charged, how does running through this gel allow us to separate them based on their size?

The gel is a matrix; which essentially means that the gel has lots of tiny pores similar to a filter. This structure allows small DNA fragments to fit through the pores, trapping the larger ones.

Larger fragments stay closer to the wells on the negative end whilst smaller fragments filter through towards the positive pole.



The dashes (-) as shown in the image above, are referred to as **BANDS**.

Bands contain a large number of DNA fragments of the same size, which have ultimately travelled to the same position in the gel.

---

## Applications of PCR + Gel Electrophoresis

Most applications involve amplifying DNA using PCR, and then loading the DNA into an electrophoresis gel to visualise the results.

**DNA Sequencing:** Process of determining base sequence of a DNA sample

1. Use PCR to amplify DNA
2. Use sanger reaction (is a method for determining the nucleotide sequence of DNA) to produce fragments for sequencing
3. Run fragments on electrophoresis gel to determine sequence

**DNA Profiling:** Process of analysing DNA variations, for the purpose of identification

1. Use PCR to amplify short tandem repeats (STRs)  
→ STR length varies between individuals, so that they can be used to construct unique DNA profiles
2. Run fragments on electrophoresis gel to determine the sizes of the STR fragments

**Recombinant DNA:** DNA which contains genes from two or more different sources

1. PCR can be used to amplify a target gene
2. Run recombinant DNA on electrophoresis gel to determine whether gene has been successfully inserted

# DNA sequencing

## What is DNA sequencing?

Process of determining the sequence of nucleotides (A, G, T, C) in a piece of DNA.

The scale of DNA sequencing varies as it can be used to determine the sequence of a single gene or a whole genome.

## Steps in DNA Sequencing:

### Gel Electrophoresis

- Collect a DNA sample
- Extract DNA from sample - obtained through chemicals which break open the cells and separates the DNA from other cell components
- Amplify DNA - using PCR to make more copies of DNA
- Perform Sanger sequencing reaction
  - goal: separately identify the position of each nucleotide
  - use 4 special PCRs = normal PCRs with a **chain-terminating nucleotide** (stops DNA synthesis)
- Determine the DNA sequence - run all 4 PCR reactions on an electrophoresis gel, to determine the lengths of the fragments in each section

### RULES FOR DETERMINING A DNA SEQUENCE

- 1 Work from **smallest to largest fragment** (one at a time).
- 2 Identify **which PCR reaction** the fragment came from.
- 3 The next nucleotide is **complementary** to the **chain-terminating nucleotide** in that reaction.

## Applications of DNA Sequencing

### Genetic Testing:

- In medicine, DNA sequencing can be used to determine if a patient is at risk of a genetic disease.
  - Genetic diseases are usually associated with the presence of particular genes.
  - An example of a genetic disease that can be identified using DNA sequencing is sickle cell anaemia.

### Biological Research:

- DNA sequencing is a useful tool in scientific research because it can be used to study genomes and the proteins they encode, at a molecular level.
  - In genome mapping, DNA sequencing is used to determine the locations of genes and the distances between them.
  - Potential drug targets and variations in drug targets can be identified using DNA sequencing. This provides an opportunity for personalised medicine.

### Evidence of Evolution:

- DNA sequencing is used in evolutionary biology to determine inheritance patterns.
  - Evolutionary biology is the study of how different organisms are related and how they evolved.

### Personal Identification:

- The uniqueness of a person's genetic code means that we can identify them by sequencing their DNA.
  - This can be used to determine parentage or the identity of a culprit/victim involved in a crime.



# DNA Profiling

## What is DNA Profiling?

DNA profiling refers to the process of analysing DNA variations, for the purpose of identification. DNA variations are specifically referred to as Genetic Markers, which are regions of DNA that usually vary between individuals.

→ Used to construct DNA profiles

- Short tandem repeats (STRs) - a string of repeating nucleotide units that are usually 2-5 bases long.
  - A string of repeating nucleotide units, where the number of units varied between people
  - Result: people have STRs that are different lengths → DNA profiles are based on these differences
- Single nucleotide polymorphisms (SNPs)
- Restriction fragment length polymorphisms (RFLPs)

## Steps in DNA Profiling:

### Gel electrophoresis

1. Collect DNA sample
2. Extract DNA from sample
3. Amplify STR fragments
  - involves using PCR to make copies of <13 different STR regions
  - this is achieved by using primers which flank the region by binding to the DNA on each side of the STR
4. Determine the length of STRs
  - amplified STR fragments are separated by gel electrophoresis
5. Interpreting the electrophoresis gel
  - patterns of bands on the gel creates a DNA profile
  - can determine the number of repeats in the STR by referring to the DNA ladder

## Applications of DNA Profiling:

### Determining Parentage (determining biological parents)

- Every person has two alleles (copies) of every STR in their gene (one from mother and other from father)
- All of the STR alleles in a person's DNA profile should also be present in their mother's and/ or father's DNA profile

### Identification

- DNA profiles can be used to determine the identity of an unknown person
  - Forensic Investigations: determines the identity of the suspect or victim
  - Natural Disasters: determines the identity of people killed in natural disasters
- A sample taken from the scene is used to generate a DNA profile.  
The victim/ suspect is the person whose DNA profile is a complete match.

## MODULE 6 - MUTATIONS

### Genetic change:

A mutation refers to a change in the base sequence of an organism's DNA.

It can be caused by mutagens (environmental agent that damaged DNA) or a cellular error (cell that makes a mistake during cell division)

- Mutagen: radiation, naturally occurring toxins and chemicals
  - Cellular error: chromosomal and point mutation

## The effects of mutation

- Mutations matter because of the effects it has on the organism
  - Mutations can occur in somatic or germ-line cells and can occur during coding or non-coding regions

## Mutagens

Mutagen - is an agent which can change the structure of DNA, causing a mutation

## Electromagnetic radiation

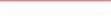
- Short wavelengths and high energy waves have the potential to cause mutations (eg, gamma + x-rays)
  - When electromagnetic waves move through matter, they give energy to the atoms they hit. Atoms react to the high levels of energy which cause them to vibrate and lose electrons. This causes the breaking of chemical bonds in the DNA. The cell dies or lives with mutated DNA
  - Sources of EMR - infrared, visible and UV light. Radioactive elements (eg uranium 236)
    - X-ray machines, PET scanners

## Chemicals

- Mutations occur when chemicals
    - are accidentally incorporated into DNA instead of proper nucleotides
    - Inserts itself into DNA
    - Chemicals can make gaps in the DNA

## Naturally occurring mutagens

- Mutagenic chemicals which specifically come from microbes, plants and animals

Radiation	Chemicals	Infectious Agents
 UV (from sunlight)	 X-rays (medical uses)	 Carcinogens (e.g. cigarettes)

# Point Mutations

Point mutations: a change in only one nucleotide. Point mutation occur due to spontaneous error occurring during DNA replication

- **INSERTION** - extra nucleotide is added
- **DELETION** - a nucleotide is not included
  - Both insertion and deletion of nucleotides in DNA are called **FRAMESHIFT POINT MUTATIONS**
  - This is because when a nucleotide is added/ removed, the entire chain of nucleotides shifts, changing the triple codon order and purpose
  - This can have a major effect since it changes the amino acid sequence
- **SUBSTITUTION** - wrong nucleotide is added
  - When a single base is substituted, the mutation is missense, nonsense or silent
  - Missense - substitution of one nucleotide, so the codon codes for a different amino acid
  - Nonsense - substitution of one nucleotide, so the codon is a stop codon. This prematurely stops the growth of the correct polypeptide
  - Silent - The wrong nucleotide is substituted in, but the tRNA still codes for the same amino acid. This does not affect the function of the amino acid

MUTATION	POINT
DEFINITION	Only <b>one nucleotide</b> is changed
CAUSES	Error in DNA replication
PROCESSES	Frameshift (insertion & deletion) or substitution (mis-sense, nonsense & silent)
EFFECTS	<b>Frameshift:</b> all codons after mutation are affected (= different amino acids) <b>Mis-sense:</b> different amino acid <b>Non-sense:</b> unfinished protein <b>Silent:</b> no effect
EXAMPLE	<b>Sickle cell anaemia:</b> point mutation in haemoglobin gene (GAG → GTG)

**EXAMPLE** - Sickle cell anemia: a genetic disorder where people have misshapen red blood cells. This is caused by the substitution of a single nucleotide of a gene that creates haemoglobin. The normal code if GAG but the subbed code if GTG; this changed the amino acid from GLU to VAL which causes haemoglobin to fold into an abnormal shape, clumping together

# Chromosomal Mutations

Chromosomal Mutations: A change in the arrangement or structure of a chromosome

Caused by spontaneous errors in cell division. Crossing over increases the probability of mutations

- Deletion - a section breaks off and is lost
- Inversion - a section breaks off, flips around and reattaches
- Translocation - a section breaks off and sticks to a different chromosome
- Duplication - a section is accidentally copied more than once
- Non-disjunction - chromosomes don't separate properly

In all chromosomes, if breakage occurs in the middle of a gene, the gene will be destroyed

In deletion, inversion and translocation mutations, genes are moved to a new place

In duplication, the end result will change the amount of proteins produced

Effects of nondisjunction is not as distinct

CHROMOSOMAL
Change in structure of <b>chromosome</b>
Error in cell division
Deletion, inversion, translocation, duplication & non-disjunctions
<b>All:</b> breakage in middle of gene destroys the gene <b>Deletion, inversion &amp; translocation:</b> genes in another place <b>Duplication:</b> changes in amount <b>Non-disjunction:</b> varied effect
<b>Trisomy 21:</b> non-disjunction during gamete formation (3 #21)

EXAMPLE- (nondisjunction) down syndrome (trisomy 21) is a genetic disorder where people have three copies of chromosome 21 instead of two.

## Germ-line and Somatic Mutations

Somatic cells are normal body cells and germ cells or gametes are sex cells

A germ-line mutation is a change in the DNA of a germ cell

- Effect- has no effect in the organism that it occurs in but all mutated cells will be passed on to all offspring

Somatic mutations are changes in the DNA of a somatic cell

- Somatic cells have nothing to do with the passing of genes to the next gen
- Effects - a somatic mutation will only affect the daughter cells of the mutated somatic cell. Offspring is unaffected, unlike germline mutations when mutations affect offspring

MUTATION	GERM-LINE	SOMATIC
DEFINITION	Change in DNA of <b>germ cell</b> = a cell which forms sex cells	Change in DNA of <b>somatic cell</b> = a body cell
EFFECT (ON ORGANISM)	<b>None</b>	All <b>daughter cells</b> from the original, mutated cell will have the mutation
EFFECT (ON OFFSPRING)	All <b>cells</b> will have the mutation	<b>None</b>
EXAMPLE	Down syndrome	Lung cancer

## Mutations in Coding and non-coding regions

Coding DNA : DNA which codes for the amino acid sequence of a protein

- The effect if a mutation in coding DNA depends on the type of mutation

Non-coding DNA : DNA that does not code for proteins.

Non-coding DNA has three functions; it makes functional RNA, regulatory sequences and repetitive sequences. If a mutation occurs during these functions...

- Makes functional RNA - protein synthesis is affected
- Regulatory sequence - Gene transcription levels may change. Even though the gene looks normal, the amount of proteins can increase/ decrease
- Repetitive sequences - if mutations occur, nothing happens, no impact

MUTATION	CODING DNA	NON-CODING DNA
DEFINITION	DNA which codes for the amino acid sequence of a protein	DNA which doesn't code for proteins
EFFECT OF MUTATION	Vary depending on type of mutation and what cell it's in	Makes functional RNA: change in efficiency of protein synthesis Regulatory sequences: change in amount of protein produced Repetitive sequences: none
EXAMPLE	Sickle cell anaemia	Lung cancer

# Gene Pools

The order of bases in DNA acts like an instruction manual for how to make all of the proteins in a cell. DNA can be separated into smaller segments called genes which code for specific features of the organism. Alleles are different versions of the same gene eg. eye colour

**Definition** - Gene pools are total collections of alleles for all genes in a population. We can use different gene pools to compare different populations. Gene pools are dynamic - they change over time due to change in allele frequency.

## Factors that affect gene pools

**GENE FLOW** - Movement of alleles between populations, due to movement of individual organisms. If gene flow between populations are high, then their gene pools are going to be similar. If gene flow between populations are low or absent, then their gene pool are going to be become increasingly different (can be so different they cannot breed anymore)

**GENETIC DRIFT** - Random events occurring within the population can lead to changes in the gene pool. These genes in the gene pool of the population are the genes of the 'lucky' individuals, not necessarily the 'better' individuals.

Example: a person steps on a bunch of brown beetles and leaves a higher population of green beetles alive. By random events, the beetle population has a gene pool with a high frequency of green alleles.

**MUTATIONS** - A change in an organism's DNA. Mutations are a source of new alleles, so that the size of the gene pool is increased. This mutation introduces an entirely new allele into the population, increasing the genetic diversity/ gene pool of a population.

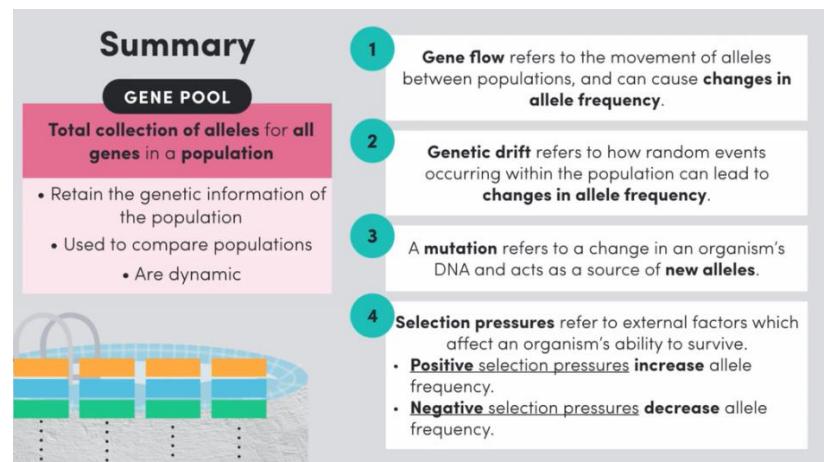
**SELECTION PRESSURES** - Any external factor which affects the organism's ability to survive in its environment. Selection pressures change the allele frequency in a population, as organisms with advantageous alleles are more likely to survive

Can be influenced by:

- Weather
- Temperature
- Predators
- Food
- Mates

**Positive selection pressure** increases the frequency of an allele in a gene pool

**Negative selection pressure** decreases the frequency of an allele in a gene pool



# Biotechnology

Biotechnology is the use of biological systems, processes and organisms in the creation of new products and technologies. The goal of new biotechnologies is to improve the quality of human life. Biotechnology is used in many different fields; however, its most significant role is in medicine, agriculture and industry.

Historical examples of biotechnology include:

1. Selective breeding in agriculture - This deliberate selection and breeding of organisms with the most favourable characteristics is known as selective breeding, and it is the earliest example of biotechnology. Overall, the goal of selective breeding is to retain favourable characteristics in future generations.
2. Fermentation
3. Traditional medicine

## Uses and Applications of Biotechnology in the Present

### Polymerase Chain Reaction (PCR)

Used to amplify a specific region of DNA. This is particularly important since scientists often need large amounts of DNA if they are to analyse, test or use it.

### DNA Sequencing

Where the sequence of nucleotides (A, G, C, T) in a piece of DNA is determined.

### DNA Profiling

Where a person's DNA profile is created by analysing DNA regions which are usually different between different people.

### Cloning

Where a genetically identical copy of a gene, tissue or organism is made.

### Transgenic Organisms

Which contain genes taken from another species.

## KEY NOTES:

- Biotechnology is the use of biological systems, processes and organisms in the creation of new products and technologies.
- Modern biotechnology primarily involves molecular techniques which enable us to manipulate DNA, as well as improvements to historical biotechnology.
- Modern uses and applications of biotechnology include PCR, DNA sequencing, DNA profiling, cloning and transgenic organisms.
- In the future, it's likely that biotechnology will play a key role in medicine, agriculture and industry.
- When it comes to deciding how and when to use biotechnology, evaluating the implications of our decisions on the social, ethical and economic aspects of humanity is crucial.

# Reproductive Technologies

## Types of Reproductive Technologies

### Artificial Insemination

Artificial insemination involves deliberately introducing male sperm into the female reproductive tract, by a method other than sexual intercourse. The process involves collecting sperm from a male, which may be used immediately or may be frozen and stored before being inserted into the reproductive tract of a fertile female.

### Artificial Pollination

Artificial pollination is basically just the plant version of artificial insemination. That is, it refers to when humans manually facilitate the natural plant pollination process. This involves the transfer of pollen from the stamen (male part) of one flower to the stigma (female part) of another flower.

There are two methods for performing artificial pollination:

- Mechanical pollination- large amounts of pollen are released from aeroplanes or blowers directly onto the plants. Although this is relatively quick and doesn't require much labour, not many flowers are successfully pollinated, meaning it's pretty inaccurate.
- Hand pollination - a person uses a small brush to transfer pollen between flowers. This is obviously super labour intensive, but it has a much higher accuracy rate than mechanical pollination.

## Positive Outcomes of Reproductive Technologies

Artificial insemination and artificial pollination can be used to positive effect in:

- Producing offspring with desirable characteristics.
- Overcoming geographical barriers between organisms.
- Improving the rate of reproductive success.
- Offspring with Desirable Characteristics

## Overcoming Geographical Barriers

The ability to transport semen and pollen over large distances means that genes can be spread across the world, increasing biodiversity. This is particularly crucial to wildlife conservation projects where it may not be possible to transport individuals over long distances.

## Improving Reproduction Rates

A good example of this is how artificial insemination can be used in humans to improve the chances of a successful pregnancy. This technology is used when a male or female has fertility issues or if donor sperm is being used.

Artificial pollination can quickly achieve the same results as natural pollination, which ultimately reduces harvest times.

## Negative Outcomes of Reproductive Technologies

When used excessively, artificial insemination and pollination can lead to reduced biodiversity since only a few individuals are allowed to contribute to the next generation. Over time, "desirable" traits become more and more common, whilst "undesirable" traits are bred out.

This means that the population is:

- Less likely to survive sudden environmental changes, and
- More likely to suffer from inbreeding.

- Reduced Resilience to Sudden Changes

## Inbreeding

With increased inbreeding, it's more likely that less favourable genes (such as those which are responsible for recessive genetic diseases) will show up more frequently in the phenotype.

# Whole Organism Cloning

**Definition:** The process of making an exact genetic copy of a whole organism

## Cloning Whole Plants:

### Cutting

1. A section is removed from the parent plant and placed in soil or water
2. Cutting develops its own roots, stems, and leaves
3. Cutting develops into a full-sized version of a original plant

### Grafting

1. A cutting from the stem of the plant is bonus to the cut stem of another plant with developed roots
2. Stems fuse
3. Grafting grows as if it is a part of the original plant

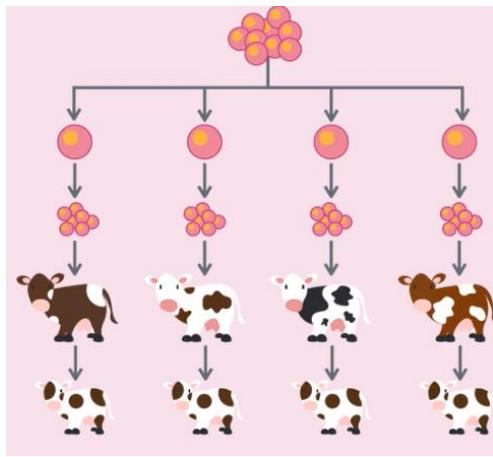
### Tissue Culture

1. A section of the parent plant is pulverised, releasing individual plant cells
2. Cells are grown on a nutrient and hormone containing medium
3. Cells grow to form small sprouts
4. Sprouts are moved onto another medium to grow further, or are re-introduced to natural growing environment

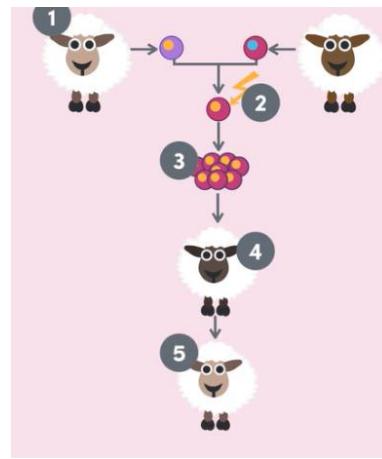
## Cloning Whole Animals:

### Artificial Embryo Twinning

1. Egg is fertilised by sperm, forming a cell
2. Cell develops, forming a clump of identical, unspecialised cells → each of these cells can develop into a complete organism
3. Identical cells are split, forming embryos
4. Surrogate mothers give birth to genetically identical offspring



Artificial Embryo Twinning



SCNT

### Somatic Cell Nuclear Transfer (SCNT)

1. A donor cell is taken from the organism to be cloned, and an unfertilised egg is taken from a female organism
2. DNA from egg is removed and replaced with DNA from the donor cell
3. Egg cell is triggered to divide by an electrical impulse, developing into an embryo
4. Embryo is transplanted to surrogate mother
5. Surrogate mother gives birth to genetically identical copy of organism

### Applications of Whole Organism Cloning:

#### Scientific Research

- Animal testing plays a key role in the development of drugs and medical devices
- Cloned animals produce reliable responses to drugs used in research
- Theoretically, these cloned animals have the same reactions to the same drugs

#### Agriculture

- Cloning allows desirable traits to be passed onto offspring
- Faster and more reliable than natural breeding
  - (Eg. tissue cultures enable the quick production of lots of genetically identical plants)

#### Conservation of Organisms / Wildlife Conservation

- Cloning can be used to prevent the extinction of critically endangered species
  - (Eg. White rhinoceros)
- Cloning may be used to bring back species from extinction
  - (Eg. use of woolly mammoth DNA)

### Exam Application:

*"Artificial selection and animal cloning are similar biotechnologies, used to create organisms with improved characteristics."*

Evaluate this statement. In your response, include reference to the processes involved in the two biotechnologies. (7 marks)

Definitions

Similarities

**Improved Characteristics**

While selective breeding is focused on developing improved characteristics in farm animals, manipulations to select desirable characteristics, such as increased animal product yields, can have adverse effects on the organism. For example, breeding the Jersey cow and Friesian bull generates issues of painful udders due to the large quantities of milk production.

On the other hand, animal cloning is not focused on creating improved characteristics, but rather on keeping the next generation identical. Cloned animals also experience shorter lives than non-modified organisms and hence characteristics are not improved. For example, a cloned sheep called Dolly lived for 6.5 years despite the species' 12 year life expectancy. This demonstrates that the statement is not true overall.

# Gene Cloning

Gene cloning is the process of making an exact copy of a particular gene

## Methods of Gene Cloning:

IN VITRO (in a test tube)

- Polymerase Chain Reaction (PCR)
- PCR is a technique used to make lots of copies of a specific region of DNA
- 3 Steps that involve - Denaturation (95deg c), Annealing (55deg c), Extension (72deg c)

IN VIVO (in a living organism)

- Recombinant DNA - DNA which contains genes from two or more different sources

## Applications of Gene Cloning:

Direct Applications - applications interested in the SNA of the gene itself

- DNA sequencing
  - Gene cloning can make enough gene copies for scientists to perform analysis that determines the base sequence of a DNA molecule
- DNA profiling
  - Scientists can generate DNA 'fingerprints' using DNA produced in gene cloning

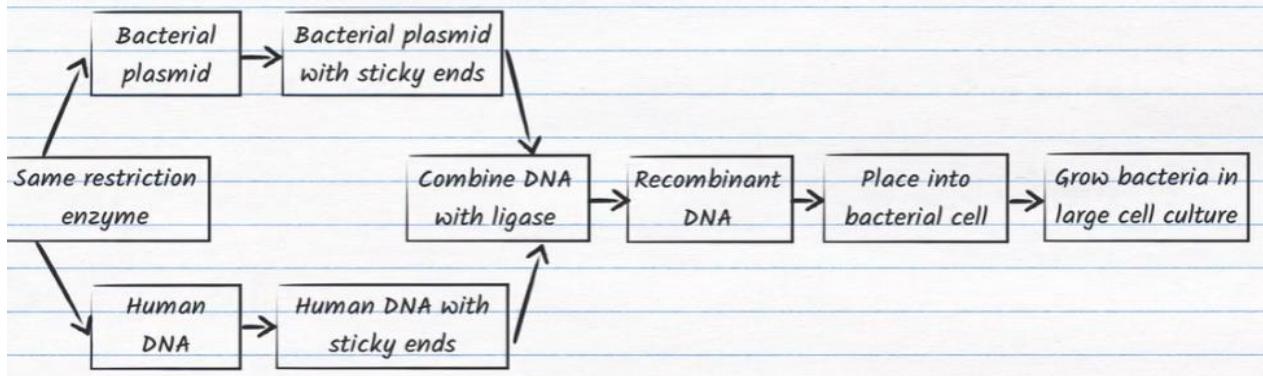
Indirect Applications - applications involving a cloned gene being inserted into an organism

- Gene Analysis
  - Genes can be added or removed from organisms as a part of research
  - Allows understanding of how a gene works
- Gene therapy
  - Scientists replace a disease-causing gene with a normal gene
  - Prevents or corrects expression of genes resulting in genetic diseases

## Exam Application:

Construct a flow chart to summarise the process of 'gene cloning' in a named example. (4 marks)

Cloning the human insulin gene in *E. coli*



# Exam Applications of Cloning

## Defining “Investigate” and “Assess”

- ★ “Investigate” means to “plan, inquire into and draw conclusions about”
- ★ “Assess” means to “make a judgement of value, quality, outcomes, results or size”.

So basically, for this topic, NESA wants us to be able to say whether we think whole organism cloning and gene cloning effectively achieve their goals, taking their advantages and disadvantages into consideration.

## Whole Organism Cloning:

Whole organism cloning is the process of making an exact genetic copy of a whole organism.

→ It can refer to making cuttings, graftings and tissue cultures in plants, as well as artificial embryo twinning and somatic cell nuclear transfer in animals. It has lots of different goals, depending on the application.

ADVANTAGES	DISADVANTAGES
<p>Scientific research</p> <ul style="list-style-type: none"><li>• Cloning can produce genetically uniform animals that produce reliable drug responses.</li><li>• Cloning can be used to help save human lives by enabling the production of "grown-to-order" tissues (embryonic stem cell technology).</li></ul>	<p>Extremely costly/ expensive process</p> <ul style="list-style-type: none"><li>• Hard access to right equipment</li><li>• Requires lots of time and money to produce cloned products</li></ul>
<p>Agriculture</p> <ul style="list-style-type: none"><li>• Cloning can be used to produce crops and livestock with certain “desirable” characteristics.</li><li>• Cloned organisms can also be produced in a short time period, compared to normal breeding techniques.</li><li>• Genetic uniformity in crops results in more consistent requirements, growth rates and harvesting times.</li></ul>	<p>Ethical questions</p> <ul style="list-style-type: none"><li>• Cloned animals tend to suffer more from adverse health issues and have higher mortality rates</li><li>• Unknown health effects on humans due to cloned foods</li></ul> <p>Reduced Genetic Diversity</p> <ul style="list-style-type: none"><li>• The population is less likely to survive sudden environmental changes and is more likely to suffer from interbreeding.</li></ul>
<p>Wildlife Conservation</p> <ul style="list-style-type: none"><li>• Cloning can be used to save critically endangered species. As of yet, this hasn't been applied for animals, but has been used to save rare plants (like the Wollemi pine) from extinction.</li></ul>	

## Example of a conclusion:

Taking the above points into consideration, I believe that whole organism cloning is a valuable and effective technique to produce genetically identical organisms to be used in scientific research/agriculture/wildlife conservation. My judgement stands true, so long as the long-term risks associated with cloning are managed appropriately. The decision of whether to use whole organism cloning must be considered on a case-by-case

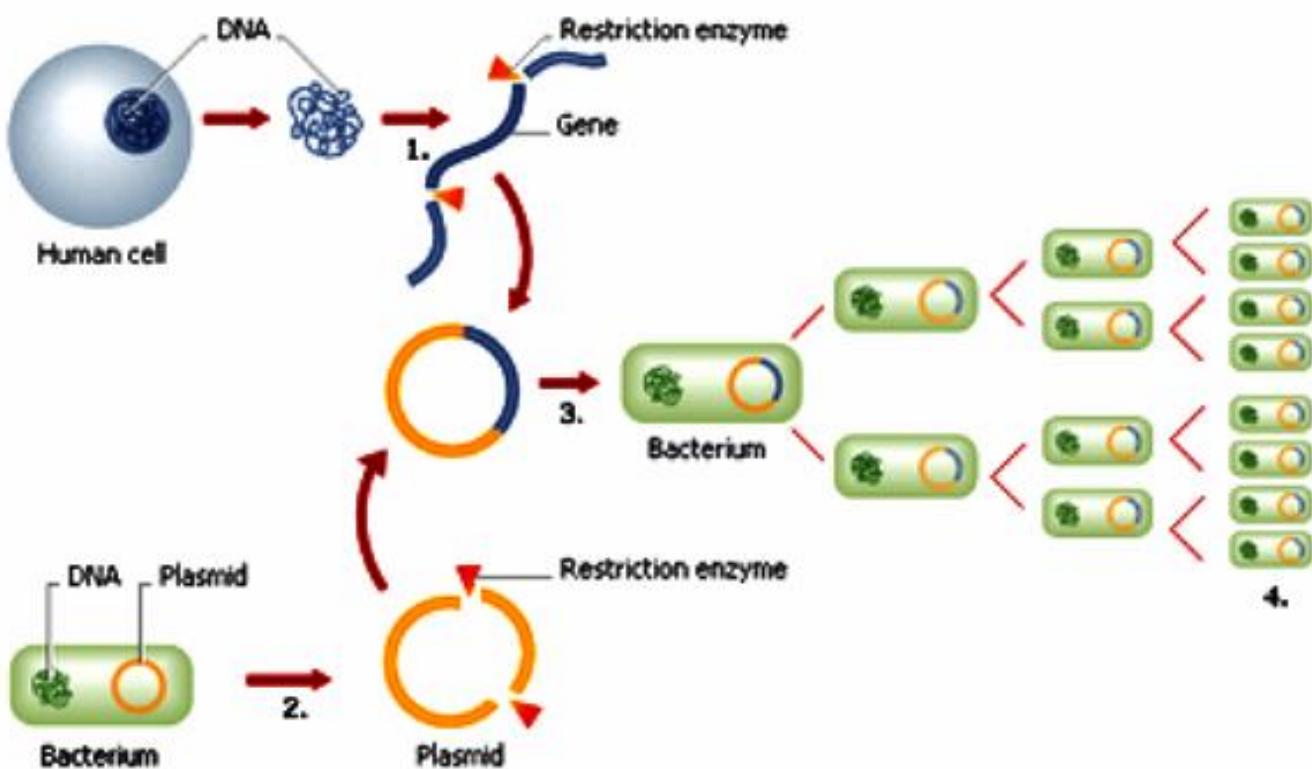
basis, in order to accurately judge whether the potential benefit outweighs the risks and to implement strategies to optimise benefits.

## Gene Cloning

Gene cloning refers to the process of making an exact copy of a particular gene.

→ The two main forms of gene cloning are *in vitro*, using a polymerase chain reaction, and *in vivo*, using recombinant DNA technology and bacterial transformation. Again, gene cloning has different goals, depending on the application.

ADVANTAGES	DISADVANTAGES
<p>DNA sequencing and DNA profiling</p> <ul style="list-style-type: none"><li>Gene cloning is crucial to ensure there is a large enough sample for analysis. → This is important, since we may only have a small DNA sample available.</li></ul> <p>Take forensic investigations, for example. Merely touching an object, like a door handle or some car keys, doesn't leave much cellular material for analysis. However, this DNA can be amplified to larger amounts by gene cloning so that we can identify the individual responsible for the crime!</p>	<p>Expensive process</p> <p>Ethical questions</p> <ul style="list-style-type: none"><li>The use of gene cloning for DNA sequencing and profiling doesn't bring up too many ethical issues generally, so long as the DNA is collected with a person's consent and privacy is upheld.</li><li>However, the ethical issues surrounding transgenic organisms are fairly extensive.</li></ul> <p>For example, genetically modified foods may have detrimental effects on human health. Again, we look at these issues in more depth at another time.</p>



# Recombinant DNA

All living organisms use DNA to store their genetic code → the genetic code is universal

- Scientists can transfer genes from one organism to another, so that the organism to which the gene has been transferred to is able to express the genes to make proteins.

**Recombinant DNA** - DNA which contains genes from two or more sources

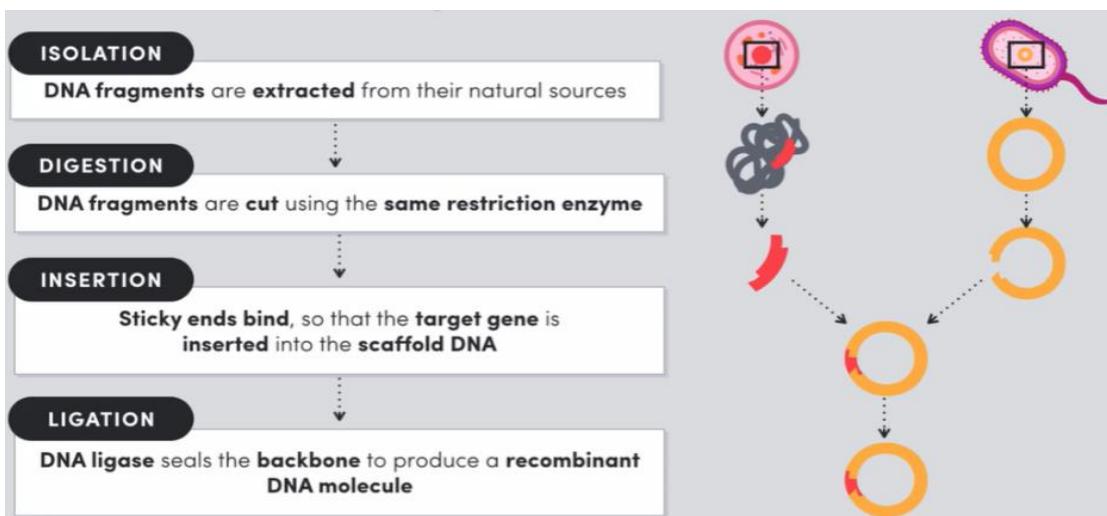
## Making Recombinant DNA:

1. Isolation - DNA fragments are extracted from their natural sources
  - Target Gene - Gene we are interested in, because of the protein it produces
  - Scaffold DNA - DNA molecule that the target gene is going to be inserted into

### Plasmids

- Most common scaffold used in recombinant DNA technology
- = **small, circular** piece of DNA
- Found in **bacteria**
- Usually code for **genes which aren't crucial** to the functioning of the organism, e.g. antibiotic resistance
- Naturally **transferred between bacterial species (vector)**

2. Digestion - Digestion refers to something that is being broken down. In this step, DNA fragments are cut using the same restriction enzyme (which cuts DNA at a specific base sequence)
  - Each restriction enzyme cuts at a specific base sequence of the target gene
  - The enzymes are also used to cut two ends of a plasmid. The exposed bases are referred to as sticky ends which are complementary to the target gene
3. Insertion - Sticky ends bind, so that the target gene is inserted into the scaffold DNA
  - When the target gene and plasmid DNA are connected, the ends spontaneously stick together to form a recombinant plasmid
  - the exposed base pairs recognise a complementary sequence and causes it to quickly match up according to the base, officially forming a recombinant plasmid
  - Even though the bases have bound, the backbone is still broken
4. Ligation - DNA ligase seals the backbone to produce a recombinant DNA molecule



# Bacterial Transformation

Bacterial transformation is the process of introducing foreign DNA into bacteria.

→ To amplify the target gene, we introduce the recombinant plasmid into a bacterial cell. This is because it is easy to grow lots of bacteria, and consequently, lots of target genes.

1. Combine plasmids with bacteria
  - Add both components to a solution containing calcium ions
  - Calcium ions disrupt the bacterial cell membrane, allowing recombinant DNA to move into the cell through the membrane
2. Heat shock the bacteria
  - Involves quickly increasing the temperature, for a short amount of time
  - Forces recombinant DNA into the cell
  - Since the recombinant plasmid is being used to transport the foreign gene into bacteria, we refer to it as a 'vector'
3. Allow bacteria to recover
  - Bacteria are transferred into nutrient-rich broth, at their optimal temperature (~37 deg celsius)

## Selecting Transformations:

To select the transformants, scientists use plasmids with a selectable marker

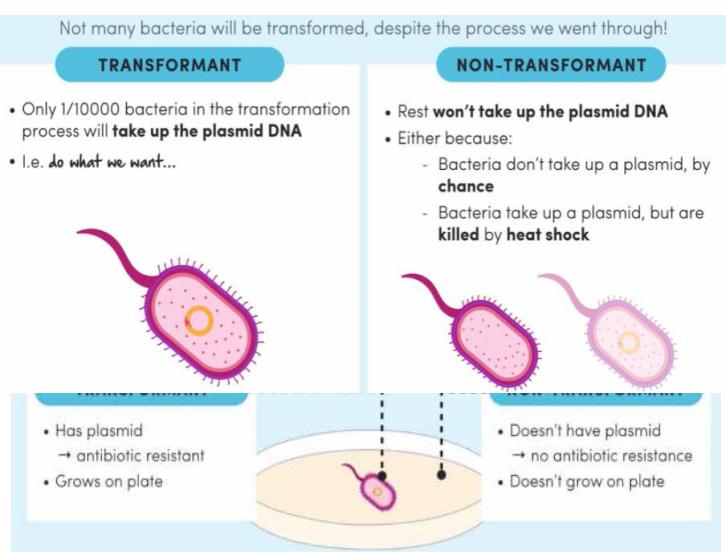
→ selectable markers are genes that produce easily observable characteristics

→ usually, the selectable marker is an antibiotic resistance gene which allows bacteria to survive in the presence of a specific antibiotic

→ To figure out if a bacterium has a plasmid, the bacteria is plated onto agar plates containing the antibiotic.

## Uses of Transformants:

- Main reason is to make multiple copies of the target gene
  - This is done by amplifying the gene, resulting in a large culture of the transformed bacteria
  - Eg. placing bacteria into nutrient-rich broth at their optimal temperature
- "Protein Factory"
  - Uses transformed bacteria as "factories" to make the protein (encoded by the target gene)
  - Protein is then harvested and purified  
→ Eg. insulin to treat diabetes
- "Plasmid Factory"
  - Recombinant DNA is extracted from the transformed bacteria
  - Copies of the target gene are used for other things  
→ Eg. DNA sequencing, DNA profiling, making transgenic organisms



# Making Transgenic Organisms

Biotechnology: Using biological processes, systems and organisms for the benefit of humanity

Genetic Engineering: The process of modifying the genome of a living organism

Genetically Modified Organisms (GMOs): The products of genetic engineering. It may involve removing a gene, adding a gene or altering a gene

Transgenic Organism: GMOs which have been modified to contain genes from another species

Transgenes: The gene that is transferred from one species into another

→ All transgenic organisms are GMOs, but not all GMOs are transgenic organisms

→ The genetic code is universal. Meaning, almost any gene transferred from one organism to another will express the protein expressed in the original organism.

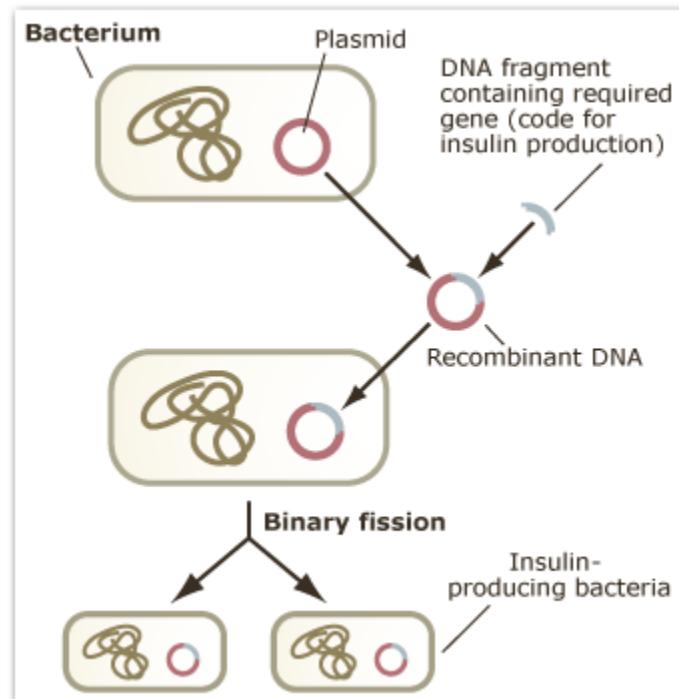
→ Simply, genes from one organism will still work the same way when it is put into another organism

→ The production of transgenic organisms is made possible by recombinant DNA technology

## Techniques for Making Transgenic Organisms:

- aka how genes are inserted in the DNA of another organism

1. Bacterial Plasmids: A small circular piece of DNA used to transfer genes to other species. Recombinant plasmids are a great option to transfer DNA information.
2. Microinjection: DNA from one species is inserted into a cell from another species using a micropipette.
3. Biolistics: A gene gun shoots small metal bullets coated in DNA into the nucleus of a cell.
4. Electroporation: cells are quickly exposed to a series of short electrical impulses, creating a hole in the cell membrane through which DNA can move into the cell. This is useful for inserting larger DNA fragments when other techniques cannot make a hole big enough.
5. Viruses: viruses naturally insert genetic material into their host cell to replicate and survive. By introducing a target gene onto the viral genome, a virus can be used to insert a gene into an organism.



# Reasons for Transgenic Organisms

## Increase Crop and Livestock Resistance:

- Increased protection against pests, diseases, and extreme environmental conditions

Eg. BT cotton contains a gene taken from soil bacterium. This gene codes for a toxin that disrupts the digestive system of cotton-eating caterpillars, killing them. Through this, farmers achieve higher yield without the need for increased insecticides; saving their time and money. Additionally, through this technology, it has a lower environmental and human health impact since pesticides are not being used excessively.

## Increase Productivity, Yield, and Quality of Crops and Livestock:

Eg. Golden Rice contains 2 additional genes from daffodils and soil bacterium which produces grains high in vitamin A, which is essential in the human diet.

Golden rice has a higher quality nutrient content → solves health problems related to vitamin A deficiencies in developing countries.

## Produce Therapeutic Products:

- Used to produce proteins including hormones, cytokines, enzymes, vaccines and antibodies which have applications in research and medicine.

Eg. Insulin is needed to treat people with diabetes. Human genes for insulin can be inserted into a plasmid used to transform E.Coli. The E.Coli then produces human insulin which can be harvested and purified.

## Study Human Diseases:

- Can be used to predict how certain drugs and treatments affect humans with certain conditions

Eg. OncoMouse contains a human cancer gene. The gene increases the likelihood of the mouse developing cancer and is then used to assist research and the development of cures for human cancer.

## Create New Products:

Eg. Blue roses - roses are modified with pansy genes to express the blue colour.

Discuss ONE ethical issue arising from the use of transgenic species. (4 marks)

*Transgenic species can be produced using recombinant DNA technology, which involves the transfer of genes from one organism to another. A debated ethical issue involving transgenic species is animal welfare.*

### Advantages

*Recombinant DNA technology has provided the ability to produce proteins previously extracted from animals. For example, large amounts of human insulin can now be produced using bacterial plasmids, removing the need to extract pig insulin.*

### Disadvantages

*However, the detriment to species during medical research needs to be considered. For example, the Oncomouse is genetically modified to develop human cancer within the first few weeks of life. Because of this, they have a much shorter life span than regular mice, at only eight weeks.*

# The Effect of Biotechnology on Biodiversity

Biodiversity refers to the variety of life on Earth, both within and between species.

## **Conservation of Biodiversity:**

### Within a Species

Artificial insemination and artificial pollination: The ability to transport semen and pollen over large distances means that genes can be spread across the world, increasing biodiversity. This is particularly crucial to wildlife conservation projects where it may not be possible to transport individuals over long distances.

### Between Species

Biodiversity can also be increased by moving genes between species, using recombinant DNA technology.

→ Remember: a transgenic organism refers to an organism with genes which were taken from another species and inserted into its genome.

For example, blue roses are simply normal roses that have some extra genes from pansies to make them blue. By manually transferring genes between species, we create more varied organisms and increase biodiversity.

## **Loss of Biodiversity:**

Primarily, a species may become more similar as a result of efforts to produce plants and animals with more desirable traits. Additionally, there is also the possibility that GM plants and animals might escape into the wild, where they may out-compete non-GMOs, or interbreed with closely related species to produce hybrids with disastrous consequences.

### Breeding for Desirable Traits

When used excessively, artificial insemination and pollination can lead to reduced biodiversity since only a few individuals are allowed to contribute to the next generation. Over time, "desirable" traits become more and more common, whilst "undesirable" traits are bred out. This contributes to reduced biodiversity.

→ A decrease in biodiversity is concerning because it means that future generations of crops and livestock are less likely to survive sudden environmental changes, such as exposure to new pests and increased temperature.

## **Out-Competing Non-GMOs:**

If a GMO manages to escape into the wild, its impact will depend on how well it is able to survive and compete against other organisms naturally found within the ecosystem. So if a GMO is at a significant advantage, it will out-compete other organisms, leading to a loss of biodiversity.

→ Another point here is that GMOs may not just have effects on organisms occupying the same niche in an ecosystem. A loss of biodiversity can also occur due to effects on non-target organisms such as bees, beetles and moths, which either cannot feed on the GM crops or are killed by insecticides within the crops.

## **Interbreeding to Produce Hybrids:**

It is also possible that GMOs might interbreed with wild relatives to produce hybrids. If these hybrids have a significant survival advantage, they may outcompete other organisms so that biodiversity is lost.

When a pesticide-resistant GM crop cross-pollinates with a wild plant species, it's possible that a hybridised pest species called a "super-weed" might be created. That is, a plant species which we are unable to control since the resistance genes render all of our pesticides ineffective. These super-weeds can be devastating to both natural ecosystems and farms.

# Social Considerations of Biotechnology

Social considerations are those which address the implications of biotechnology on human society. They specifically concern the rights that societies protect. Everyone living in society should enjoy equal rights and protection, and this, in turn, protects society itself.

The potential risks associated with the use of products and services created by biotechnology must be assessed and evaluated, before they are released to consumers. Even if the product is deemed “safe”, any concerns must be well-advertised. For example, it’s important that GM foods are properly labelled since people have the right to know what they’re eating and make their own decisions.

Society has a role in protecting a person’s privacy, and this is executed by implementing laws. Laws to protect privacy might achieve this by ensuring a person has consented to the collection and storage of their genetic information, or by preventing discrimination based on genetic information. Because the field of biotechnology is advancing so quickly, it’s important that these laws are frequently reviewed in response to new findings, in order to ensure privacy is protected.

## Key Takeaways

- Social considerations are those which address the implications of biotechnology on human society.
  - They specifically concern the rights that societies protect.
  - Everyone living in society should enjoy equal rights and protection, and this, in turn, protects society itself.
- Social status and financial standing can impact on access to biotechnological products and services, which are often very expensive.
  - This creates social inequity since those who benefit from these products and services are those who can afford them, not those who are most in need.
  - Golden Rice - high levels of vitamin A. Vitamin A deficiency is common in developing countries where it contributes to high rates of childhood blindness and infectious disease. Although the consumption of Golden Rice could help fix the vitamin A deficiency problem, high production and distribution costs mean that developing countries often choose alternative solutions.
- Human health can be negatively impacted by applications of biotechnology including genetically modified foods and gene therapy.
  - Any risks need to be evaluated and well-advertised.
- Privacy has become a debated issue in biotechnology because many of its applications (such as DNA profiling and genetic sequencing) require the storage of people’s genetic information in databases.
  - Without proper legislation, this genetic information could be misused.

# Ethical Considerations of Biotechnology

Ethical considerations are more personal: within a society, different people will have different interpretations of aspects of biotechnology, and we need to consider each of these perspectives in our decision-making processes.

The main ethical considerations surrounding the use of biotechnology include:

- The role of a person's philosophical, cultural and religious views,
- The importance of consent in a medical setting,
- Legal implications,
- Animal welfare

## Key Takeaways

- An "ethical" consideration refers to a sensitive issue of what is morally right and wrong, and which people often interpret differently.
- Ethical views on biotechnology can be shaped by their philosophical, cultural and religious views.
  - Applications such as genetic screening during pregnancy and the consumption of genetically modified foods can be particularly ethically sensitive to some groups and individuals.
- Informed consent is needed for all medical treatments in Australia to be deemed ethical.
  - For example, before proceeding with genetic screening, parents must be informed of the potential risks to themselves and their unborn child, and of the impact on their decision-making process, in order to give consent.
- Biotechnology has both positive and negative impacts on animal welfare.
  - Certain biotechnological applications have reduced our dependence on animals, improving animal welfare.
  - Some people perceive that other applications violate animal rights, by contributing to their pain and suffering for our benefit.

Explain how the use of transgenic crop plants may have adverse biological effects. (4 marks)

Transgenic crops are created when a gene from another organism is introduced into a crop plant, using recombinant DNA technology. This allows the crop to produce a new trait which would not naturally occur in the plant.

This can have adverse biological effects by decreasing the biodiversity within a species. Introducing the same transgenes into a plant population, such as a gene from bacterium to produce Bt cotton, will decrease variation in the gene pool and reduce biodiversity. This means that if a selection pressure arises, the likelihood of a gene being present which would confer survival is reduced.

Transgenic crops can also reduce the populations of nearby species. As some genetically modified crops produce large quantities of pollen, it can be easily blown onto the food plants of other species. For example, Bt corn is genetically modified to be toxic to caterpillars. Pollen that is food plants and is ingested can decrease butterfly populations in other areas.

Evaluate relevant ethical issues raised by the use of current biotechnology. (6 marks)

Whilst biotechnologies provide a number of benefits to society, it is important to use them with care and in consideration of ethical values.

### Issue 1: Genetic Screening

Genetic screening during pregnancy allows individuals to identify genetic abnormalities within unborn babies and evaluate whether to continue with the pregnancy. However, this is an issue as certain religious and cultural groups deem human life as sacred from conception and disagree with manipulation after that point.

### Issue 2: GMOs

Genetically modified organisms can be used to increase the quantity and quality of crop yield. For example, genetically modified tomatoes are made by inserting the "antifreeze" gene from winter flounder fish so they can grow in cold climates. However, this is often contested as some cultural/religious beliefs prohibit individuals from consuming certain foods, such as meat. This means that individuals who do not eat meat may have concerns about eating GMO tomatoes containing a flounder fish gene.

Overall, biotechnologies do provide considerable advantages to society, however, ethical issues are important to consider to ensure technology is not exploiting or harming the natural environment.

# MODULE 7 - Infectious Diseases

**Disease** - A condition that impairs the normal functioning of an organism

	INFECTIOUS	NON-INFECTIOUS
NATURE	Can be transmitted from one organism from another	Cannot be transmitted from one organism from another
CAUSE	Pathogens - any organism which is capable of causing disease	Genetic and lifestyle factors (eg. nutrition and environment)

## Types of Pathogens

### Bacteria

Prokaryotic, unicellular organisms which don't have membrane bound organelles and are each made of one cell.

#### Key defence mechanisms:

Not all bacteria can cause illnesses, however, pathogenic bacteria can cause disease usually by releasing toxins or damaging host tissues.

#### EXAMPLES

- **Tuberculosis**

Caused by the bacterium *Mycobacterium tuberculosis*  
They enter the lungs when a person inhales **infectious droplets**

- Tetanus (humans)
- Crown Gall (plants)

### Fungi

Eukaryotic, non-photosynthetic organisms with a cell wall.

Fungi can be unicellular (yeasts) or multicellular (mushrooms).

#### Key defence mechanisms:

The damage caused by fungi mostly arises from the enzymes they produce; fungi secrete digestive enzymes and chemicals into their surroundings in order to break down organic matter. Then they absorb the simple nutrients.

#### EXAMPLES

- **Athletes foot (humans)**

Caused by different fungal species;  
- *Microsporum*,  
- *Epidermophyton*  
- *Trichophyton*  
They live in the outside layer of human skin, where they produce chemicals which break down keratin  
Symptoms include **itchiness, inflammation and flaky skin**  
• *Chytridiomycosis* (frogs)  
• *Stem rust* (plants)

### Protists

Eukaryotic, unicellular microorganisms without a cell wall.

#### EXAMPLES

- **Malaria (humans)**

Caused by *Plasmodium*  
The *Plasmodium* pathogen floats freely in the blood of an infected person  
It feeds on Haemoglobin in the red blood cells causing them to pop  
Symptoms include:  
• Fever                    • Nausea                    • Weakness  
• Muscle Pain            • Headaches  
• *Phytophthora dieback* (plants)

## Viruses

Non-cellular entities, consisting of a single type of nucleic acid (DNA or RNA) encased in a protein coat (capsid).

### Key defence mechanisms:

Viruses need a host cell to reproduce. They attach themselves by penetrating to the host cells and hijacks the host's enzymes and nutrients to make its own viral proteins and nucleic acid.

These viral mechanisms assemble into new viruses which are released from the host cell and infect others.

### EXAMPLES

#### • Influenza (humans)

Caused by the Influenza virus which damages human lung tissue

Most symptoms are caused by the body's own immune response, caused by the release of cytokines from infected cells

#### Symptoms include:

• Fever      • Coughing      • Nasal Congestion

• Ross River fever (humans)

• Tobacco Mosaic virus disease (plants)

## Prions

Non-cellular infectious proteins, which are abnormally folded versions of a protein needed within an organism

### Key defence mechanisms:

Prions can lead to serious problems depending on the structure, function and location of the normal protein.

Prions usually affect brain and neural tissue, which leads to neurodegeneration.

### EXAMPLE

#### • Kuru

Caused by a prion found in contaminated human brain tissue

It is spread by eating the brains of infected individuals

Found in New Guinea

• Creutzfeldt-Jakob disease

## Macroscopic Parasites

Multicellular pathogens which can be seen with the naked eye.

### Key defence mechanisms:

These parasites tend to cause disease by competing for nutrients from the host. They can also produce toxins and damage host tissue.

### EXAMPLE



#### Ticks

#### Fleas

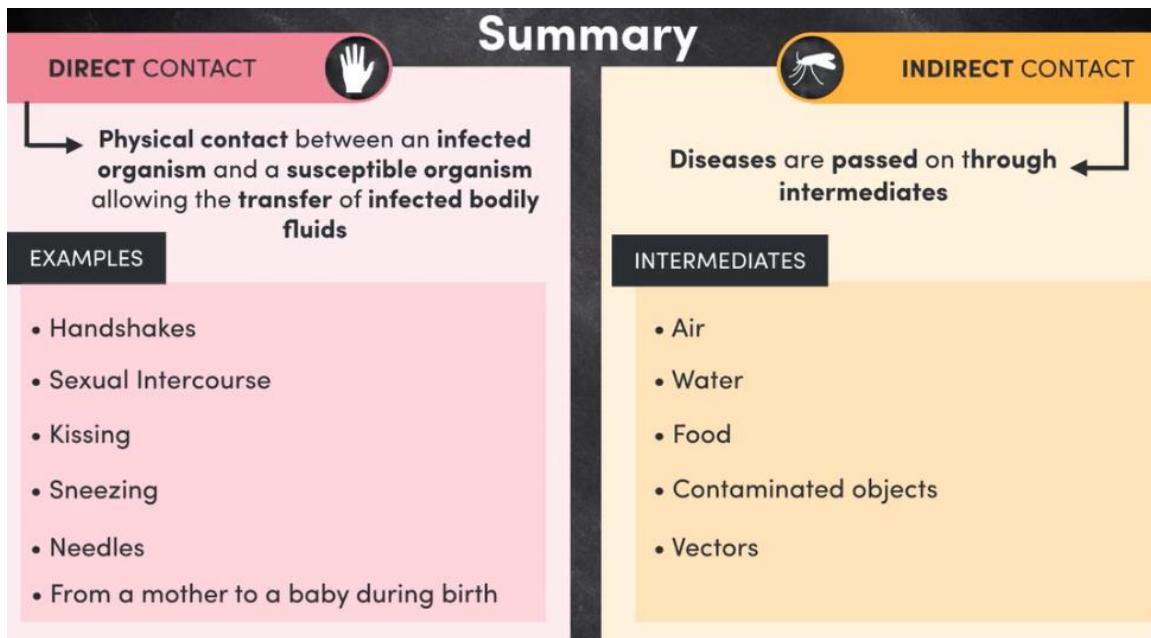
#### Tapeworm

Suck human blood to obtain nutrients

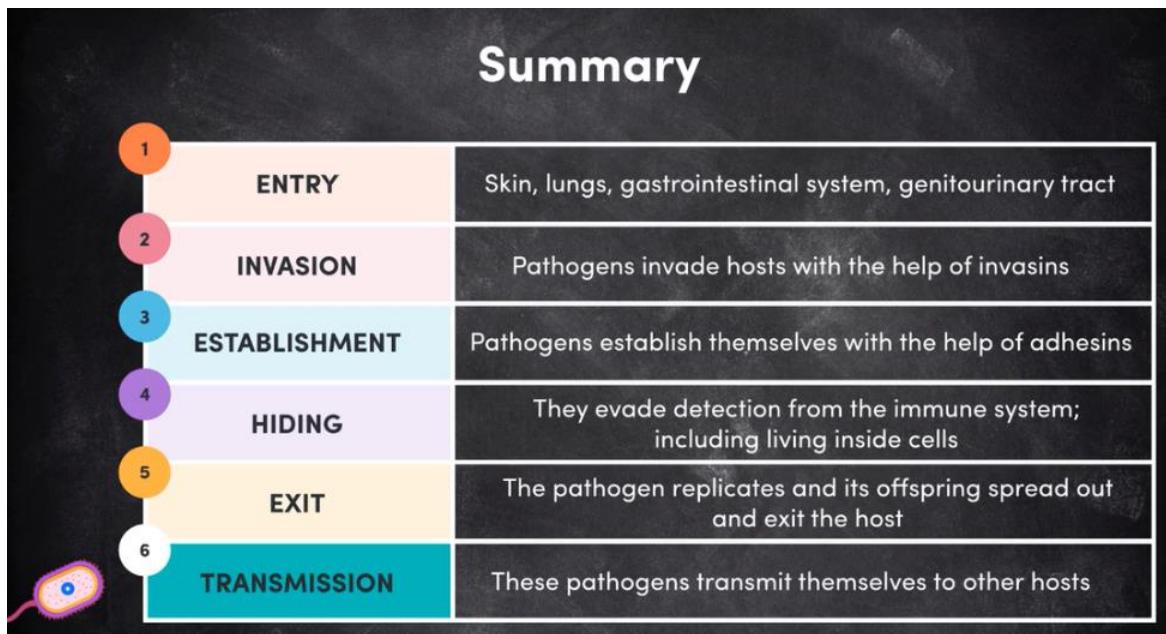
## Summary

PATHOGEN	DEFINITION	DISEASES
BACTERIA	Prokaryotic, unicellular organisms	Tuberculosis, Tetanus (humans), Crown Gall (plants)
FUNGI	Eukaryotic, non-photosynthetic organisms with a cell wall	Athletes Foot (humans), Chytridiomycosis (frogs), Stem Rust (plants)
PROTISTS	Eukaryotic, unicellular organisms without a cell wall	Malaria (humans), Phytophthora dieback (plants)
VIRUSES	Non-cellular, non-living entities	Influenza (humans), Ross River Fever (humans), Tobacco Mosaic Virus Disease (plants)
PRIONS	Infectious proteins	Kuru, Creutzfeldt-Jakob disease
MACROSCOPIC PARASITES	Multicellular pathogens which can be seen with the naked eye	Ticks, Fleas, Tapeworm

# Methods of Transmission



## How Pathogens Establish an Infection



# Epidemics

## What are epidemics?

Epidemic - an increase in the number of people affected by a disease. It involves a particular disease in a particular area. They occur when there's an increase in the number of people affected than normal.

Pandemic - an increase in the number of cases throughout a continent or across the world

Outbreak - where the disease appears temporarily in an isolated area



## Factors contributing to epidemics:

### Pathogen level

- Virulence - pathogen's ability to infect or damage a host
  - The greater the virulence, the greater effect it will have on the health of the host
  - Mutations increase a pathogen's virulence, allowing it to evade detection in hosts
- Antibiotic resistance - these pathogens can't be treated and contained, spreading easily
- Toxins - Makes hosts more susceptible to infection and it deters other pathogens from consuming food colonised by that pathogen
- Genetic shift - can occur if there is a loss of genetic variation or selection pressures that cause the population to lose genetic resistance
- Herd immunity - in populations where a significant proportion of individuals are immune, the group becomes immune

### Human level

- Migration - people can be diverse vectors, so movement between different groups increases the chances of disease being transmitted between them
- Infrastructure - when populations are densely packed together, increases exposure to pathogens is likely
  - Lack of amenities like clean water, effective sewage systems and adequate health services promote disease survival
- Healthcare - disease spread is increased where people do not have access to drugs and vaccines
  - Infrastructure is lacking, so clinics are not accessible
  - High cost of medicine

## Controlling an epidemic:

- Identifying the pathogen - Health workers help identify diseases through:
  - Clinical observation + Laboratory confirmation
  - Data is collected on the number of cases presented and fed into larger surveillance systems
- Environmental management
  - Cleansing water supplies of disease (eg. boiling, chlorinating, sealing)
  - Reducing the risk of food contamination (eg. disposing old food, limiting food preparation)
  - Creating sanitary conditions (eg. removing waste from public areas)
- Quarantine - a period of restricted movement and separation of people, animals and materials which may spread infectious disease
  - Large scale - travel bans, border regulation

- Small scale - protective clothing, isolation

## Robert Koch

### Koch Postulates

Koch's postulates are basically a list of criteria which must be met to prove that a particular organism causes a particular disease.

To prove a pathogen is connected to a disease:

1. The suspected pathogen must be present in every diseased individual, and absent in healthy individuals.
2. The suspected pathogen must be able to be isolated from the host and grown in pure culture.
3. A healthy potential host must develop the same symptoms as the original host, when inoculated with the suspected pathogen (essentially, the disease must be reproduced).
4. And, the suspected pathogen must be re-isolated from the second host and grown in pure culture, appearing identical to the original culture (essentially, the organism must be re-isolated).

Key Points:

- Robert Koch discovered, isolated and grew the bacteria responsible for causing anthrax.
  - In doing so, he built on the germ theory of disease.
- Koch made a list of criteria which must be met to prove that a particular organism causes a particular disease, which we refer to as Koch's Postulates.
  - This criteria consists of four main points which function as a checklist for identifying pathogens.
- Koch developed techniques for culturing and identifying microbes, including better dyes and solid agar media.
  - Through these techniques, he isolated and identified the causative agents of many diseases including tuberculosis and cholera, meaning they could be treated.
  - His work led to the development of the branch of medicine known as immunology

## Louis Pasteur

Key Points:

- Louis Pasteur discovered that one type of microorganism (yeast) was responsible for producing alcohol using sugars, whilst another type of microorganism (lactic acid bacteria) was responsible for contaminating wine and turning it sour.
  - This led to the idea known as microbial fermentation theory.
- Pasteur showed that heating liquids to 60-100°C for several minutes killed most microorganisms.
  - This technique is called pasteurisation.
- Pasteur proved the germ theory of disease (which states that many diseases are caused by the presence and actions of specific microorganisms within the body) using his swan-neck flask experiment.
  - In doing so, he disproved the theory of spontaneous generation.
- Pasteur discovered how to make vaccines by administering weakened versions of the pathogen to susceptible individuals.
  - He developed vaccines for chicken cholera, anthrax and rabies.

# Diseases in Agriculture

## Artificial Selection: Key Points

- Farmers artificially select individual plants and animals with desirable characteristics and allow them to reproduce so that these characteristics can be passed on to the next generation.
  - Artificial selection increases economically beneficial traits in crops.
  - However, artificial selection decreases genetic variation so that the population is more susceptible to disease.
  - As a consequence, selection must be carefully controlled and farmers must use responsible breeding practices which promote biodiversity.
- An example is the devastating Irish Potato Famine, caused by genetically identical potato crops all susceptible to the same fungal pathogen.

## Intensification: Key Points

- Intensive farming involves the use of various techniques to produce greater outputs, typically using the same amount of space or resources.
  - Intensive farming typically involves housing crops in high densities.
  - Farmers can reduce the likelihood of disease by housing animals in sufficiently sized enclosures, and growing plants with minimal pesticide and medicine use.
- An example is the neurological condition known as mad cow disease, which spread as a result of farmers using leftovers from dead animals to feed their living animals.

## Movement of People and Goods: Key Points

- Agricultural diseases can be spread through the movement of goods and people.
  - This is particularly problematic if the resident organisms are naïve to the disease.
  - To combat this, restrictions apply for the movement of goods, such as border control measures and quarantines.
- A key example is foot-and-mouth disease, especially as it impacted the British agricultural industry following the importation of infected meat.

# Responses to Pathogens + Immunity

## Introduction to Immunity:

Non-Specific (innate)	Specific (adaptive)
Repels all pathogens equally	Targets all specific pathogens
Everyone is born with non-specific defence mechanisms	It develops and adapts in response to pathogens which we encounter and fight off throughout our lives
Includes barriers to infection and includes the innate immune system	Includes the adaptive immune system

## Lines of Defence:

- Physical barriers -barriers that physically block pathogens from entering such as skin and mucus. This barrier does not 'discriminate against' different microbes since they are non-specific. They block out everything
- Innate immune system - is activated when the pathogen breaks through the first line of defence and includes phagocytosis and inflammation. Its job is to stop the pathogen from establishing itself in the body. It is also non specific.
- Adaptive Immune system - this line of defence is SPECIFIC. When a pathogen enters the body for the first time, the adaptive immune system identifies its weakness and targets them in order to remove or kill the pathogen. It determines its response based on the identity of the invading pathogen. The adaptive immune system does not kick in instantly.

Difference between 2 and 3 - The innate immune system does not produce memory cells unlike the adaptive immune system.

Physical Barriers	Innate Immune System	Adaptive Immune System
Aims to prevent pathogens from entering the body	Stops the pathogen from spreading and establishing itself in the body	Targets specific pathogens to inactivate, remove or kill the pathogen

## Antigens:

Antigens are molecules that are capable of triggering an immune response

	Self Antigens	Non-Self Antigens
Place of Production	Produced in the body	Originates outside the body
Examples	Cells, proteins and other chemicals which make up the body	Pathogens and their toxins, chemicals in insects or animal venom, proteins in food, transplanted organs and blood
Immune System Response	Recognises them as part of the organism and tolerates them	Recognises them as foreign objects and attacks them

## **Barriers to Infection:**

**Physical Barriers-** Physical barriers that prevent pathogens from entering the body

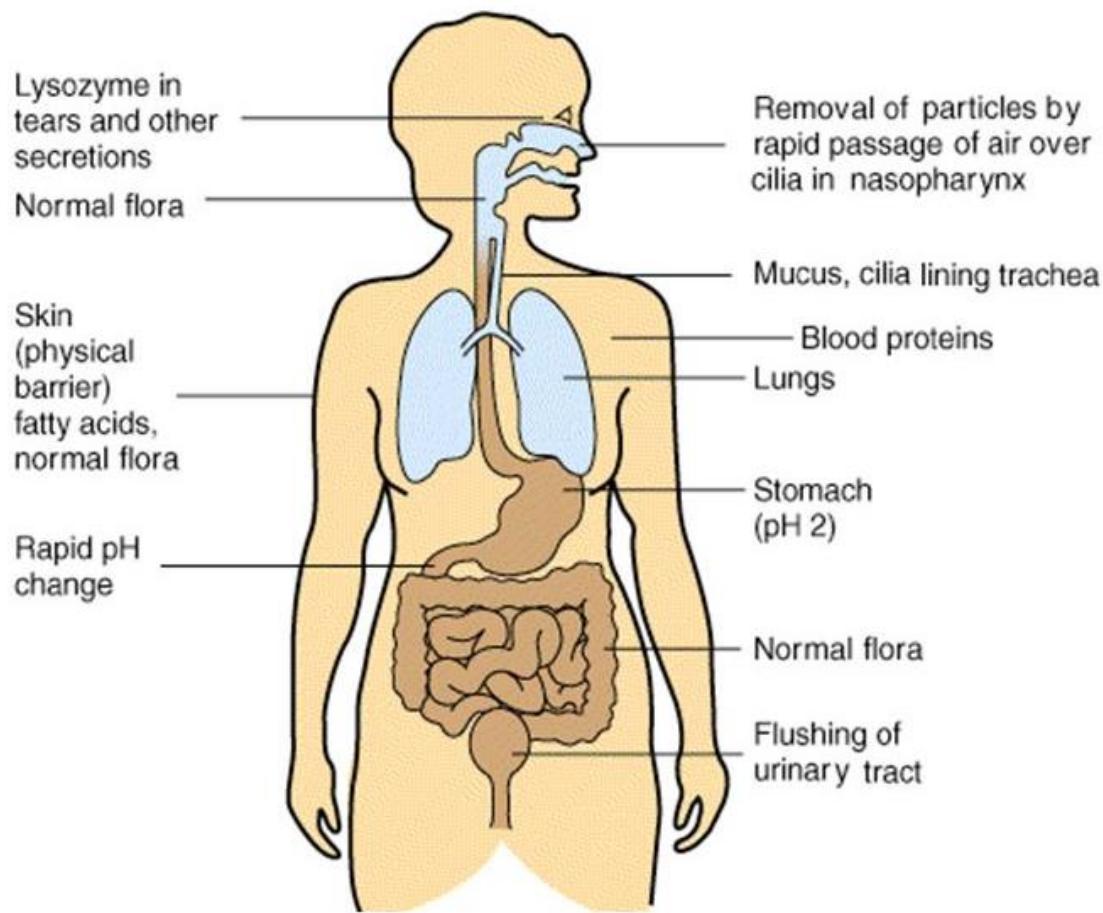
- SKIN: Tough, intact outer layer of closely packed cells. Main physical barrier against infection. Skin cells are packed with keratin which helps to strengthen the skin against breakage and infection.
- MUCUS MEMBRANES: mucus lines the openings of the body that are not covered in skin. This includes the respiratory, urinary, digestive and reproductive tracts. Mucus prevents membranes from drying out and it traps unwanted pathogens and substances until they can be removed by cilia. Mucus contains lysosomes to help break down microbes.
- CILIA: are small hair-like projections and extends from the epithelial cells which line the respiratory tract. They sweep the mucus along the tract so they can be coughed, sneezed out or swallowed.

**Chemical Barriers-** make the body surfaces inhospitable to pathogens

- LYSOZYME: An enzyme which breaks down the cell walls of bacteria, killing them and helping to fight infection. It is found in tears, saliva and mucus. The digestive enzymes in the stomach are also important.
- ACIDIC SECRETIONS (Toxic Metabolite): they contain degradative enzymes like lysozyme. Body secretions also chemically screen out pathogens by being very acidic (the secretions have a low pH)

**Microbiological Barriers-**

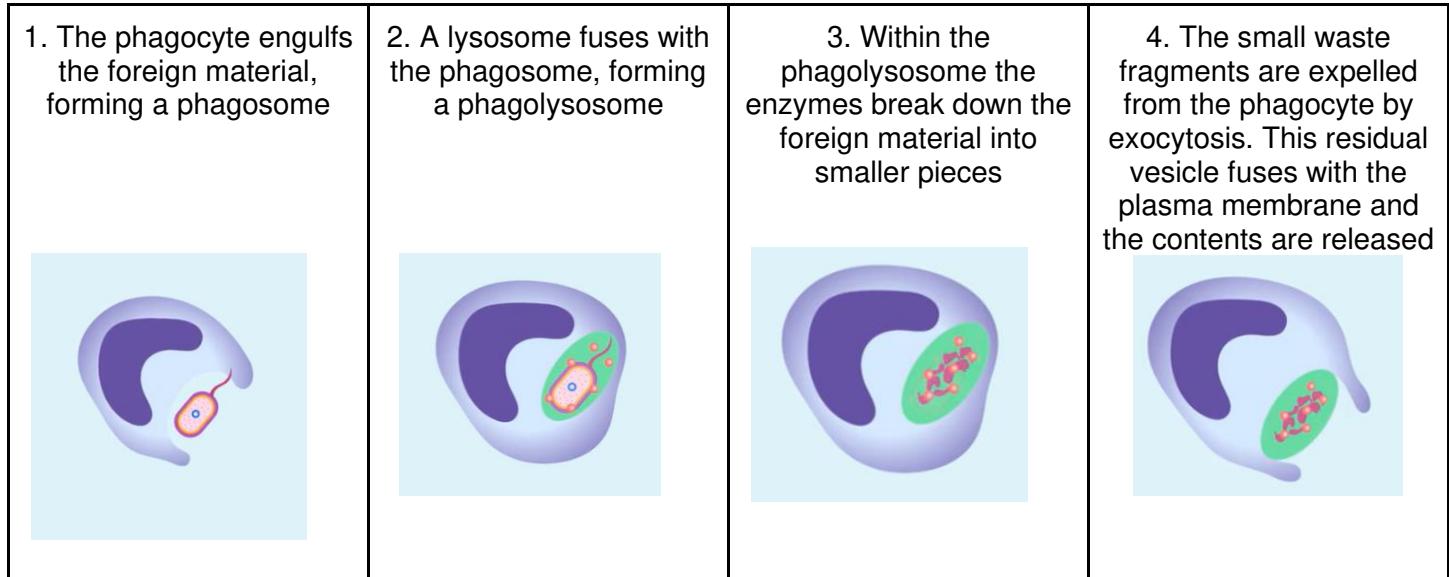
- MICROFLORA: Microorganisms in our body which don't cause harm. They are found in and on the mouth, skin and intestines. They compete for space and nutrients which makes it difficult for pathogens to establish itself into the body since it is already occupied by microflora.
- PRODUCE ANTIMICROBIAL CHEMICALS: antimicrobial chemicals prevent the growth of other organisms.



# The Innate Immune System

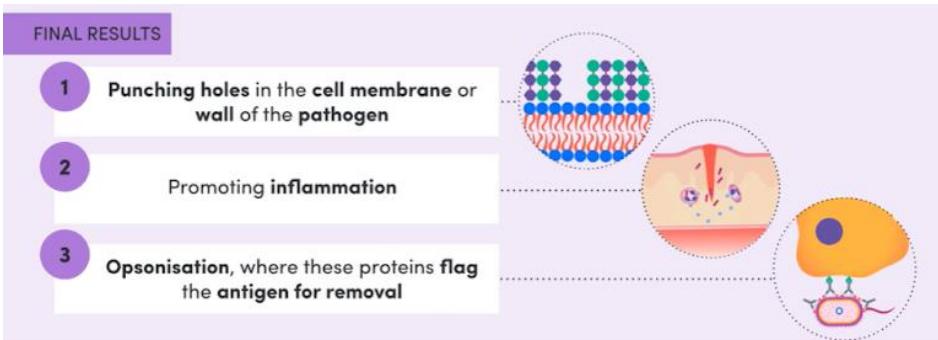
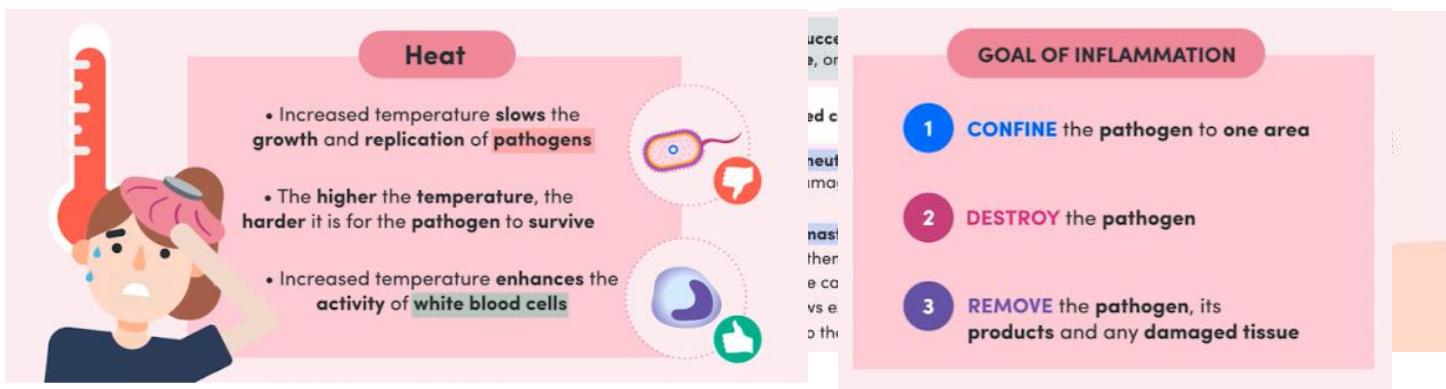
The innate immune system is responsible for non-specifically stopping foreign materials from spreading throughout the body, once they've made it through the barriers to infection

**PHAGOCYTOSIS** - phagocytosis is the process by which it engulfs and destroys foreign or unwanted material. Phagocytes are a type of white blood cells (leukocytes) and are designed to protect the body against invaders and are split into neutrophils, macrophages and mast cells.



Phagocytosis		THE INNATE IMMUNE SYSTEM		Summary
NEUTROPHILS		1	PHAGOCYTOSIS	The process by which <b>phagocytes</b> engulf and destroy foreign or unwanted material
• Phagocytosis: Quickly enter tissues and phagocytose pathogens in acute infection	• Phagocytosis: They live longer than other phagocytes so they help fight chronic infections	2	NATURAL KILLER CELLS	Target <b>virus-infected</b> and <b>cancerous</b> cells by releasing chemicals which kill them <b>directly</b> , when they are in <b>close proximity</b>
• Antimicrobial Compounds: Release hydrogen peroxide which disrupts bacterial and fungal cell membranes killing them	• Antigen Presentation: Present some antigenic fragments on their surface  It then meets up with T lymphocytes and activates them  • Release cytokines	3	INFLAMMATION	The accumulation of <b>fluid</b> , <b>plasma proteins</b> and <b>white blood cells</b> that occurs when tissue is <b>damaged</b> or <b>infected</b>
• Release cytokines Which attract other immune cells and promote inflammation		4	THE COMPLEMENT SYSTEM	Results in the <b>activation</b> of <b>proteins</b> that <b>damage</b> the <b>invading pathogens</b> , as well as <b>increased inflammation</b> and <b>activity</b> of <b>phagocytes</b> and <b>antibodies</b>

**NATURAL KILLER CELLS** - they constantly patrol the body and are important in defence against its targets of virus-infected and cancerous cells. They release cytotoxic chemicals which can kill the cell directly and they only release it when they are in close proximity to the target cell.



**INFLAMMATION** - refers to the accumulation of fluid, plasma proteins and white blood cells that occur when tissue is damaged or infected. This immune response is referred to as the Inflammatory Response.

**Cell mediated immunity** : involves the action of T Lymphocytes/ cells.

a T Lymphocyte comes into contact with a specific antigen, it proliferates and differentiates into four cell types.

T cells are made in the bone marrow and are released into the blood mature in the THYMUS GLAND.

When

to

of

#### SYMPTOMS

- Redness
- Heat
- Swelling
- Pain
- Loss of function in the affected area

#### Cytotoxic t-cells (Killer T cells)

- Kill foreign, infected and abnormal cells. They are also capable killing body's own cells
- Cytotoxic T cells secrete or inject toxic chemicals into the target cell

#### Helper t-cells

- Help promote the activities of other immune responses by secreting cytokines
- Increases the activity of phagocytosis
- Helps promote inflammation
- Stimulates the production of cytotoxic t cells
- Stimulates B Lymphocytes to differentiate into plasma and memory cells

#### LIFE CYCLE

- They are produced in the bone marrow
- They are released into the blood and mature in the thymus gland
- Once they mature, they're released into the blood again where they circulate in an inactive state
- If a T lymphocyte comes into contact with its specific antigen, the receptors on its surface allows it to bind it and the cell becomes activated

#### Suppressor t-cells

- Turns off the immune response after the antigen has been successfully contained, destroyed or removed

## **Memory T-cells**

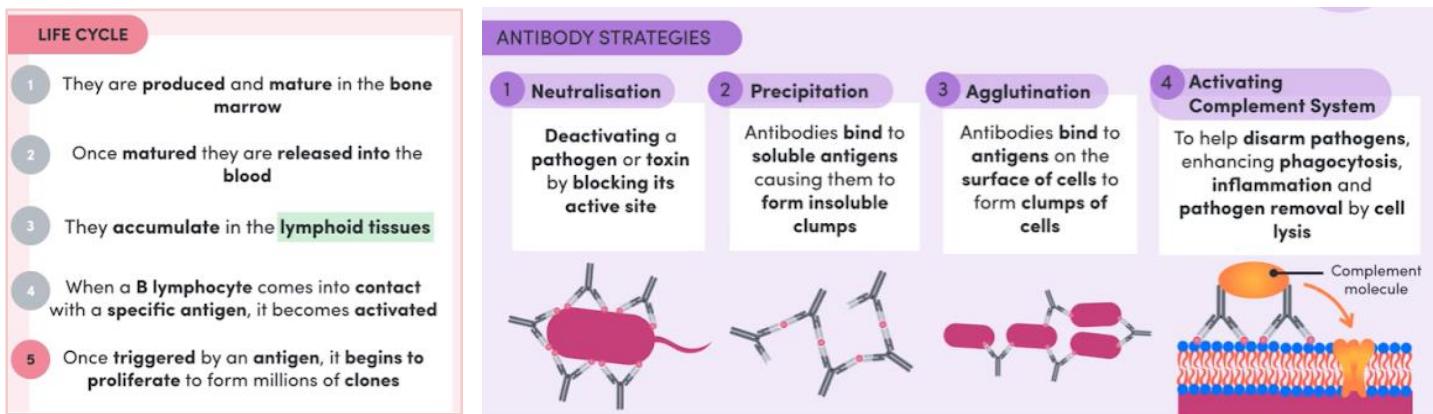
- Provides the body with long term defence against antigens
- They persist after an infection, to enable a larger and faster response upon reinfection with the same pathogen
- If the body is exposed to the same antigen, the memory t-cells will recognise it and divides into cytotoxic and helper t-cells

# Adaptive Immune System

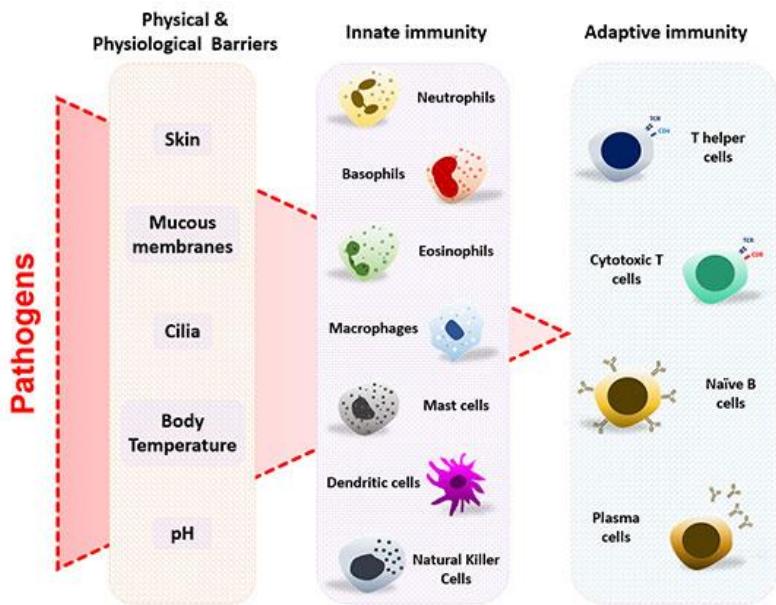
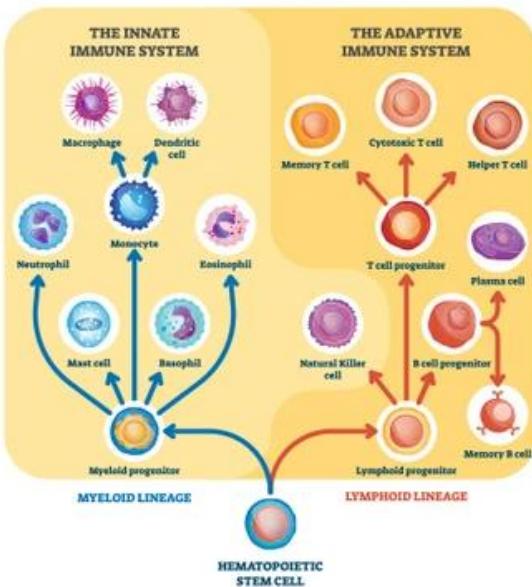
**Antibody mediated immunity:** involves B Lymphocytes which become activated and proliferate when stimulated by specific antigens.

B lymphocytes / cells is a type of white blood cell produced in the bone marrow. It differentiates into two types, plasma cells and memory B cells.

- Plasma cells - are Y-shaped proteins that produce antibodies and bind to specific antigens that trigger the B lymphocyte. When they bind together it forms an Antigen-Antibody complex
  - Antibodies interfere with the functioning of a pathogen in a way that either
    - The pathogen is unable to cause damage
    - It is easier for other components of the immune system to destroy it
- Memory B Lymphocytes - provide immunological memory and is a long term defence against antigens.
- Memory B cells lay dormant in the lymph tissue. However, If the animal is exposed to the same antigen again, memory cells recognise it and divide to produce antibody-producing plasma cells.
  - This means that the immune response to a familiar antigen is: faster, stronger and longer lasting



## CELLS OF THE IMMUNE SYSTEM

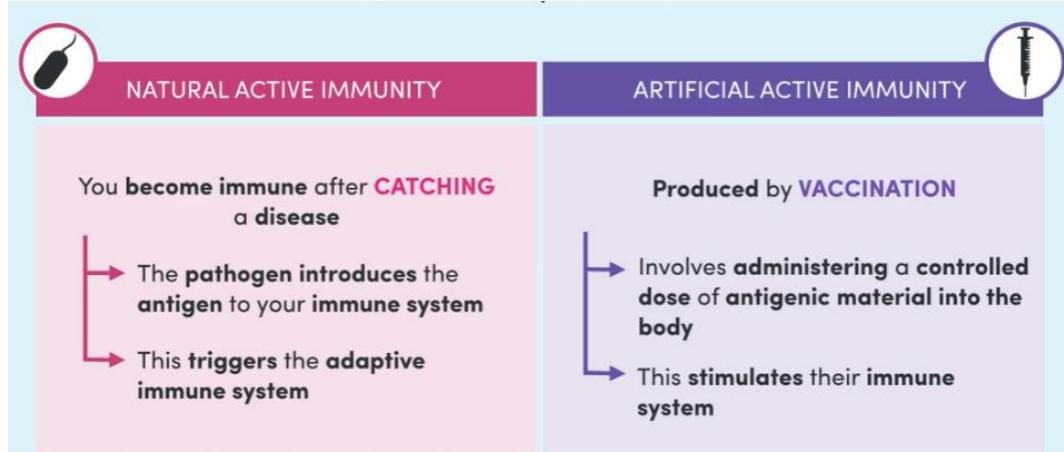


# Types of Immunity

Immune - When the body is able to protect itself without symptoms arising. The body's ability to resist infection.

**Active Immunity:** Occurs when the immune system is stimulated by an antigen to make its own T cells, B cells and antibodies. This is called 'active' because the body is proactively making itself immune.

Memory Cells = PERMANENT active immunity (constantly scans and can recognise pathogen)



**Passive Immunity:** When antibodies are transferred to an unimmunised person, providing them with temporary protection against a microbial agent or toxin. The bodies become immune after being **GIVEN** antibodies

No memory cells = TEMPORARY passive immunity



# Primary and Secondary Responses

The primary immune response happens when a pathogen enters the body for the first time. Because this is the first time the immune system has seen this pathogen, and its antigens, it can take a while for the matching T lymphocyte to be found and activated. This means there's a delay before the relevant B cells are activated, and matching antibodies produced.

As a result of how slow this whole process is, the infected person shows symptoms of the disease. This is generally fine as a one-off, but it would be rather problematic if our bodies reacted this way every time the same pathogen presented itself.

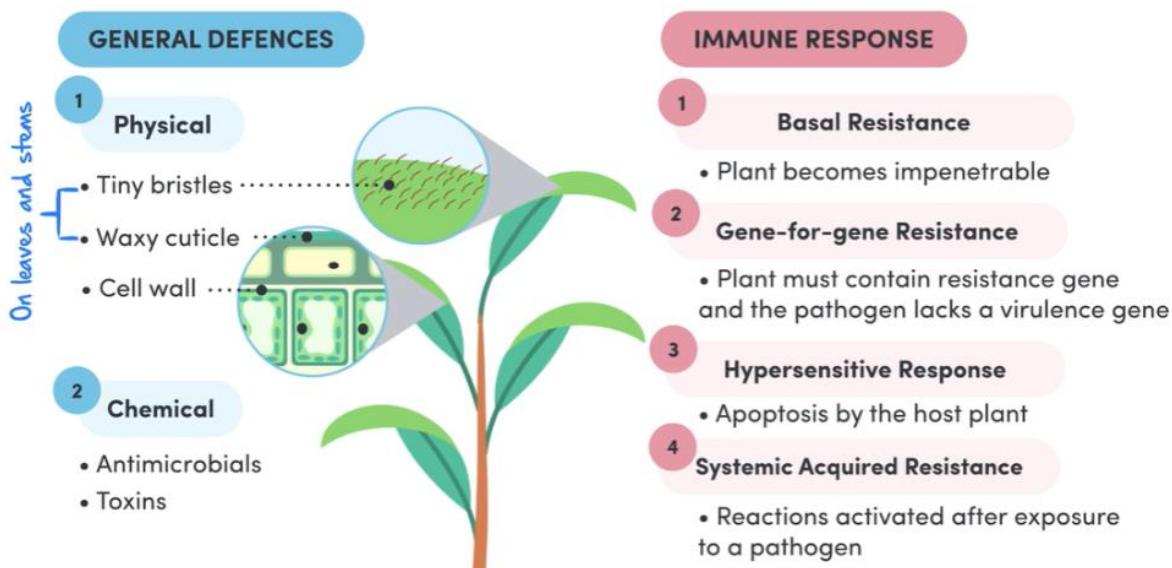
The secondary response occurs when pathogens enter the body for the second time.

It makes a difference whether the body has already encountered a specific pathogen or not, because of the T and B memory cells it produces during its first encounter. Memory cells persist after an infection, and they enable a larger and faster response upon re-infection with the same pathogen. That is, if the body is re-exposed to the same antigen, the memory cells will recognise it and divide into their corresponding cells:

- memory T cells differentiate into cytotoxic, helper and suppressor T cells
- memory B cells differentiate into plasma cells.

In this response, the whole process happens a lot quicker the second time around. Since this process is so speedy, the pathogen is destroyed before the individual even shows symptoms.

## Immunity in Plants



# Factors Influencing the Spread of Disease

## Pathogen Features

- Growth - how quickly and easily the pathogen replicates
- Survival - dictates how far the pathogen is able to travel and its opportunity to infect. It also determines the method of transmission.
  - Examples of adaptations to improve their chance of survival include the flagellum on bacteria and the protective layer on protists
- Persistence - allows pathogens to replicate over a longer period of time. They invade the host's immune system.
  - Eg. pathogens are able to lay dormant and can have slow metabolic processes

## Method of Transmission

- Can be direct or indirect
- The method of transmission affects the spread of disease since diseases are more likely to travel further via indirect transmission

## Population Density

- Diseases are spread quicker when people are in close proximity to each other
- High population densities are linked to overcrowding and lack of infrastructure

## Movement of Individuals

- When infected individuals travel, they can spread the disease to those they come into contact with
- This has a higher chance of introducing foreign diseases to new countries that would ultimately impact human health and the environment
- People who have never encountered the disease do not have immunity

## Proportion of the population that is immune

- Herd immunity - the general population is immune to the disease which prevents the disease from transmitting to susceptible individuals

## How Are Germs Transmitted?



verywell

# Preventing the Outbreak of Disease

## Hygiene Practices

- Any activity that reduces the ability for pathogens to survive and proliferate is considered as a hygiene practice
- Hygiene measures prevent the spread of diseases by creating hostile environments for pathogens by destroying them



## Vaccination

- Vaccinations specifically provide individuals with artificial active immunity (against a specific pathogen that causes a specific disease)
  - Each vaccine contains antigens against a specific pathogen
  - Once injected, these antigens trigger the immune response
  - The host develops immunity to the disease without actually catching it
  - The individual actively makes their own B cells, T cells and antibodies after being stimulated by antigens from an 'artificial' source
- Links to herd immunity

## Pesticides

- Substances which control the population of pests
- Kills the pathogenic organism responsible for the disease
- Eradicates vectors of disease such as mosquitoes
- Human benefits:
  - Protects against vector-borne diseases
  - Protects crops
  - Protects livestock
  - Defends the health of communities by sustaining food sources
- Dangers of pesticides:
  - Biomagnification - pesticide accumulates in the body tissues of organisms in the food chain in increasing amounts at higher trophic levels
  - Resistance - overuse of pesticides promotes the selection of naturally resistant strains. Due to this, stronger and larger quantities of pesticides need to be developed to have a greater effect

## Genetic Engineering

- Used to modify the genetic structure of an organism using biotechnology
- This method is used to produce plants and animals which are resistant to pests and diseases
  - To disable vectors from spreading disease
  - To modify animals to produce biomedical products

- Eg. BT cotton

## Controlling the Spread of Disease

### Quarantine

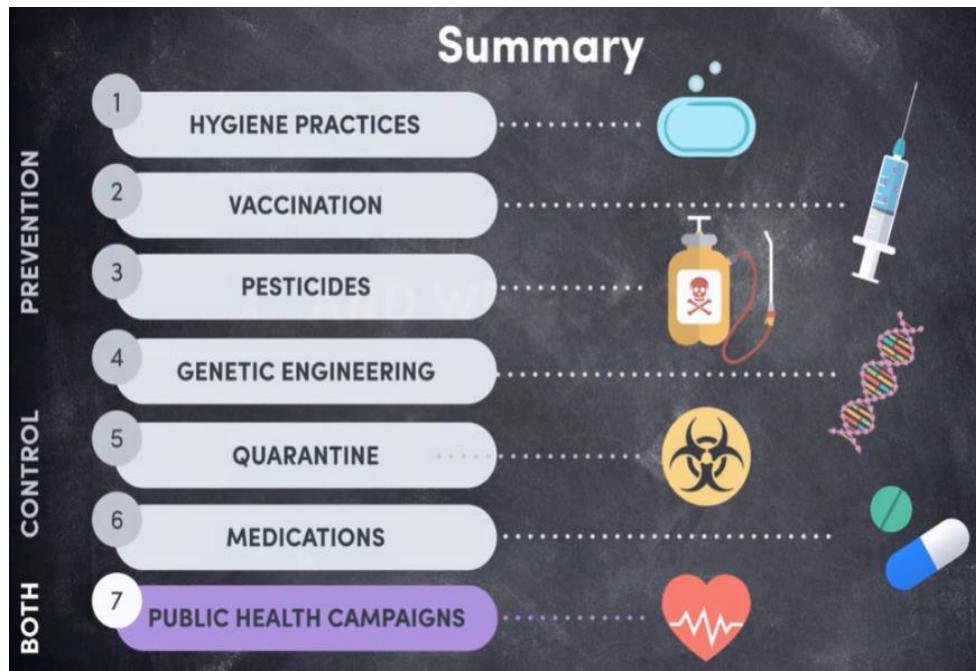
- A period of isolation used to control the spread of infectious diseases
- Quarantines only last for the period of time which the disease is communicable
- Small scale : confining patients
  - To their homes
  - To their hospital bed
- Large scale : confining whole communities
  - Borders cut off and patrolled with strict exit/entry criteria based on health
- Large scale : Goods and Transport
  - Banning the movement of goods such as fruits
  - Banning travel to highly infectious areas

### Medications

- Less restrictive method used to treat disease
- Antivirals - target viruses
- Antibiotics - targets bacteria

### Public Health Campaigns

- Used to both prevent and control disease
- There are differences between communities which are burdened by different diseases
- Campaigns vary depending on whether they are short-term intermediate response to an infection, or a long-term measure
- Promote the adoption of healthy behaviour and raises awareness of diseases



# Mosquito-borne Diseases

## The Role of Mosquitoes in Disease

Only female mosquitoes suck blood, and the reason they do this is to obtain the iron they need to grow eggs and reproduce. This means that pathogens can rely on mosquitoes to transport them to a new host.

When a mosquito takes up the blood of an infected individual, it also takes up pathogens floating in the blood. Typically, the pathogen then replicates within the mosquito and is later injected into a non-infected individual, when the mosquito bites them. At each point, the pathogen flows easily from one host to another because when mosquitoes feed they inject anticoagulants, which are substances which prevent (anti) the blood from clotting (coagulating).

In this way, the mosquito acts as a **vector**: that is, an organism that transmits disease from one host to another. Note that not all diseases are spread by mosquitoes, simply because not all pathogens are found in the blood, transferred in the blood or able to survive in a mosquito.

## Malaria

A disease caused by a protist called Plasmodium. It's carried by the female Anopheles mosquito.

When a mosquito containing Plasmodium bites a human host, the parasite enters the bloodstream, multiplies in the liver cells, and is then released back into the bloodstream. Here, it infects and destroys red blood cells. Malaria triggers fever, chills and flu-like illness. If left untreated, it can result in death.

## Dengue Fever

Dengue fever is caused by a virus called the Flavivirus, that is transmitted by the Aedes aegypti mosquitoes. Dengue is a severe, flu-like illness, which results in many symptoms including high fever, headaches, muscle and joint pains, and vomiting. Dengue fever can escalate to death through dengue haemorrhagic fever and dengue shock syndrome.

## Environmental Factors

Temperature, rainfall and humidity have a huge impact on the mosquito life cycle. They influence their rates of development, breeding and mortality. These factors play a key role in mosquito-borne diseases, as the transmission of these diseases depends on the ecology of the mosquito vector.

Excess rainfall is linked to increased transmission because it leads to the creation of natural water pools and puddles, which serve as breeding sites for the mosquitoes.

Mosquito-borne diseases typically affect the tropics, where the climate is favourable. As temperatures rise, mosquito territory increases and so does the distribution of mosquito-borne disease. And, in places already affected by mosquitoes, climate change will cause longer breeding seasons and increased hatch rates of populations. The enlarged population will cause higher rates of mosquito-borne disease.

## Social Factors

Most of the affected areas are third world countries that are **unable** to provide adequate health care, eradication methods and education programs in order to combat the spread of mosquito-borne disease. Additionally, the presence of containers of stored water, discarded junk, and dams increase the breeding grounds which are available to mosquitoes. Unplanned urbanisation also facilitates the breeding of mosquitoes for this reason.

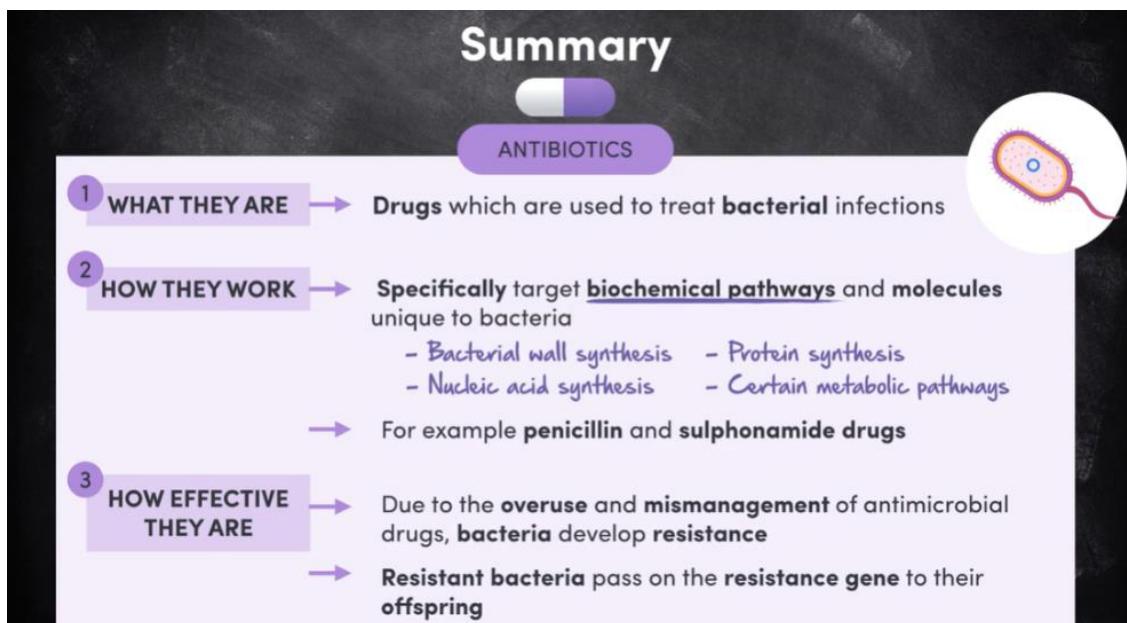
Additionally, globalisation increases the distribution of mosquito-borne disease, as individuals are exposed to new pathogens. This means that they may not have any immunity to these diseases.

# Antibiotics

- Antibiotics are drugs that treat bacterial infections by:
  - Killing bacteria causing disease
  - Slowing down the growth of bacteria, giving the immune system a greater chance of being able to remove them
- Eg. penicillin is the first antibiotic and is derived from a fungus
- Antibiotics specifically target the biochemical pathogens and molecules which are unique to bacteria
- Most antibiotics can be used to treat most bacteria
  - Not every bacteria needs a separate antibiotic since most pathogenic bacteria share the same features, allowing one antibiotic to treat a wide range of bacterial infections
  - Although, it can also kill some of the body's natural 'good' bacteria since it targets all bacteria

## Antibiotic Resistance

- Arises through spontaneous mutation
- Failure to complete antibiotic course enables resistant bacteria to survive and replicate
- Bacterial mutations:
  - deactivating/ degrading enzymes produced by antibiotics
  - Efflux pump to flush antibiotics out
  - Entry blockers to stop antibiotics to enter
- Bacteria pass on antibiotic resistance to the next generations, making the antibiotic ineffective



# Antivirals

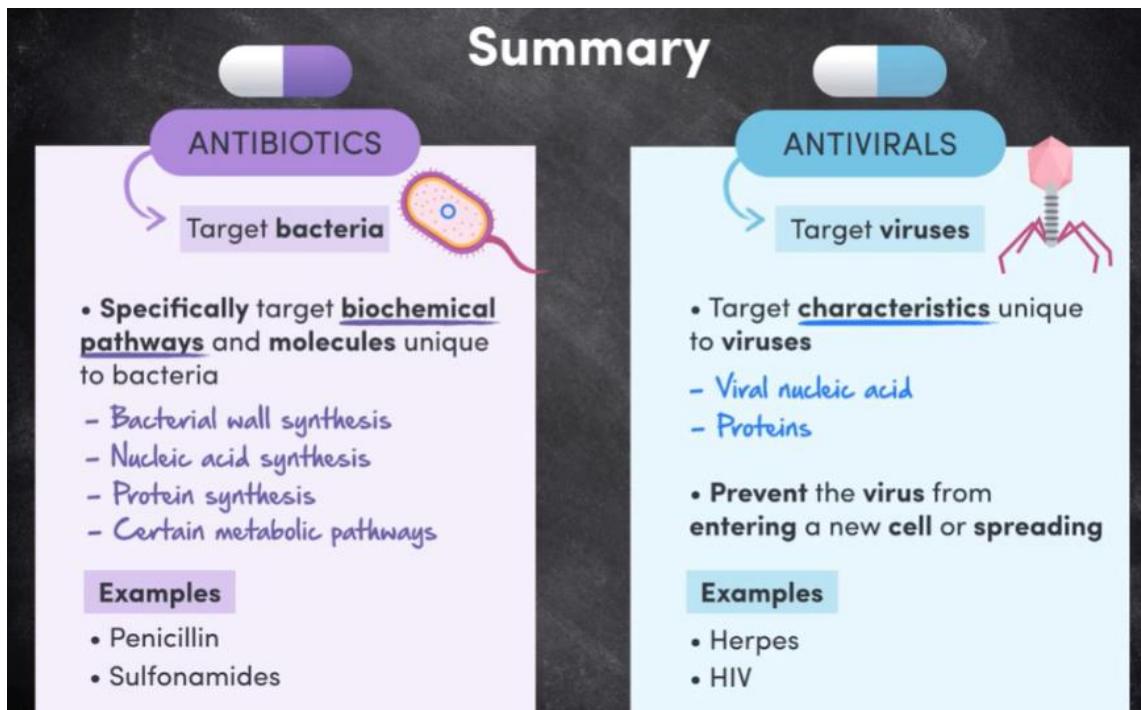
- Antivirals are drugs that treat viral infections
- They minimise the viral load by reducing the number of viruses produced in the host
- Viruses utilise the machinery of a host in order to survive and replicate
- The simple structure of viruses makes them hard to specifically target with medicine

## How they work

1. Preventing viruses from entering the host cells by binding to receptors
2. Inhibiting enzymes which catalyse the reproduction of the viral genome (prevents viral replication)
3. Blocking transcription and translation of viral proteins
  - a. Since they don't have ribosomes, viruses need to use the ribosomes of a host cell to make their protein coat
4. Preventing a virus from leaving the cell
5. Grabbing the virus whilst outside the cell by targeting capsid proteins

## How effective they are

- Viruses are difficult to target which makes antivirals slow to develop
- Some conditions may have so many viral strains that it is quite impossible to create a corresponding antiviral
- Antivirals can mutate and develop resistance
  - This is likely in hosts with compromised immune systems



# MODULE 8 - NON-INFECTIOUS DISEASES

## Homeostasis:

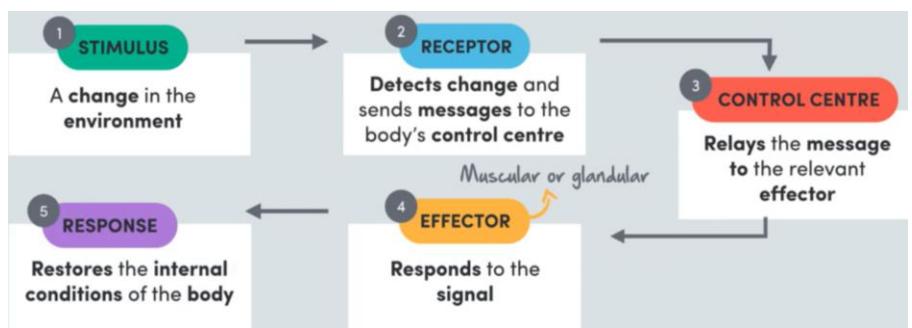
Homeostasis refers to the process by which the body maintains a constant internal environment. It uses nerves and hormones to do this. Homeostasis is important as many reactions inside the body only work under certain conditions, regardless of a changing outside environment.

### Variables:

- Body temperature
- Water availability
- Blood glucose levels
- Carbon dioxide concentration

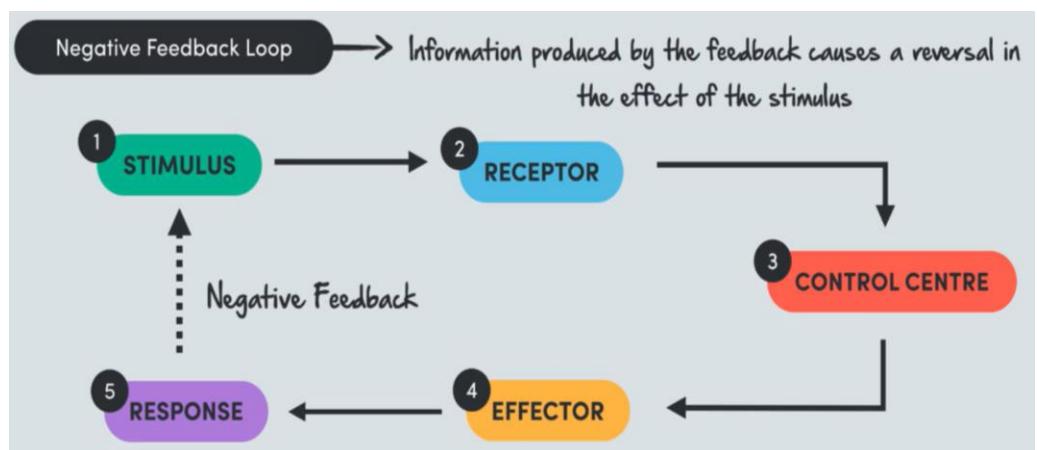
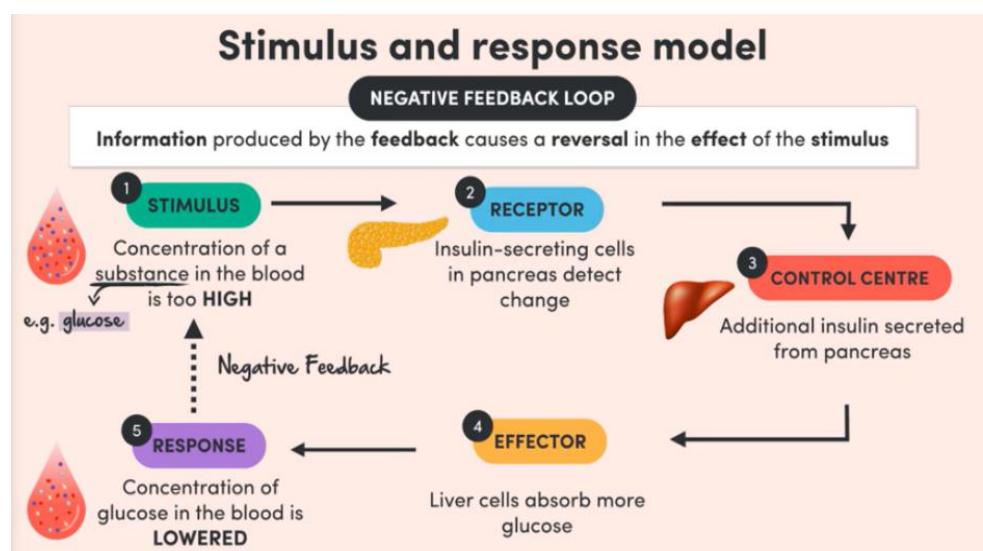
## Stimulus and Response Model:

The body's way of detecting a change in external or internal environment, and reacting accordingly.



## Negative Feedback Mechanism:

a self-regulating mechanism that maintains balance or homeostasis as a coordinated response or series of responses to a stimulus or stimuli



## **Thermoregulation:**

Refers to the internal regulation of an animal's body temperature in order to keep it within an ideal range. Humans have a preferred body temperature of 37 degrees Celsius since reactions within our body work best in this condition.

### Endotherms

- Warm blooded
- Largely create heat by adjusting processes in our body
- Relies on internal processes to maintain internal heat and homeostasis
- Rely mainly on physiological sources of heat to regulate body temperature

### Ectotherms

- Cold blooded
- Relies on external sources of temperature to heat up or cool down body temperature
- Cannot maintain homeostasis through body processes and solely depends on the environment for heat

### How it works

- Stimulus - a change in body temperature
- Receptor - the body has 2 types of receptors to detect temperature changes
  - Skin : detects external changes and are triggered frequently
  - Hypothalamus (brain) : cluster of temperature sensitive cells. Monitors the body's internal temperature by measuring the temperature of the blood
- Once these receptors detect a change, they initiate a regulatory response (can affect the amount of heat the organism generates or loses)
- Control centre - contains thermoreceptors
  - The hypothalamus is the control centre for thermoregulation
  - It receives information about temperature
  - It sends out impulses to activate the physiological response, required to re-achieve balance
- **Effectors - RESPONSES TO THE COLD**
  - Reduce heat loss
    - piloerection (goosebumps) - the response is hairs erect on skin to trap air close to the skin, preventing heat loss via convection of air
    - Vasoconstriction - blood vessels constrict, so less blood travels near the skin's surface. Less heat is lost from the blood
  - Increase heat production
    - Shivering - muscle cells perform respiration in order to break down glucose and make energy. Respiration releases heat
    - Increasing metabolism - metabolism is the main source of heat production when the body is at rest. The endocrine system can be used to influence the production of heat via metabolism
  - TSH - thyroid stimulating hormone
    - Secreted by the pituitary gland and acts on the thyroid gland to release thyroid hormones (T3 and T4 regulate metabolic processes)

- **Effectors - RESPONSES TO THE HEAT**
  - Increase heat loss
    - Vasodilation - blood actively loses heat to the external environment.
    - Sweating - this draws heat from the skin to evaporate the sweat. Converts liquid to a gas
  - Reduce heat production
    - Decreasing metabolism - in response to heat, the hypothalamus reduces the rate of cellular respiration in the body's internal organs
- Response
  - The response will try to reverse the original change in temperature
  - Eg. shivering and slowing metabolism



Pause here!

## Responding to cold

1 2 3 4

	TO REDUCE HEAT LOSS	TO INCREASE HEAT PRODUCTION
PHYSIOLOGICAL	<ul style="list-style-type: none"><li>• Goosebumps/piloerection</li><li>• Vasoconstriction</li></ul>	<ul style="list-style-type: none"><li>• Increasing metabolism</li></ul>
BEHAVIOURAL	<ul style="list-style-type: none"><li>• Seeking shelter</li><li>• Decreasing surface area</li><li>• Putting on clothes/layers</li></ul>	<ul style="list-style-type: none"><li>• Voluntary movement</li></ul>

## Responding to heat

1 2 3 4

	TO INCREASE HEAT LOSS	TO REDUCE HEAT PRODUCTION
PHYSIOLOGICAL	<ul style="list-style-type: none"><li>• Sweating</li><li>• Vasodilation</li></ul>	<ul style="list-style-type: none"><li>• Slowing metabolism</li></ul>
BEHAVIOURAL	<ul style="list-style-type: none"><li>• Covering body with water</li><li>• Swimming in cool water</li><li>• Removing clothes/layers</li><li>• Moving into shade</li></ul>	<ul style="list-style-type: none"><li>• Decreasing activity</li></ul>

# Osmoregulation

**Definition:** The movement of water from high to low water concentrations, across a semipermeable membrane

**Osmoregulation is the regulation of water to maintain homeostasis.**

Osmoreceptors in the hypothalamus detect solute concentration (osmolarity) in the blood.

## Osmoregulation + ADH

If salt concentration is too high, more ADH is released to act on the kidney, which increases water reabsorption back into the blood, and reduces urine volume.

If salt concentration is too low, less ADH is released to reduce water reabsorption, which increases urine volume.

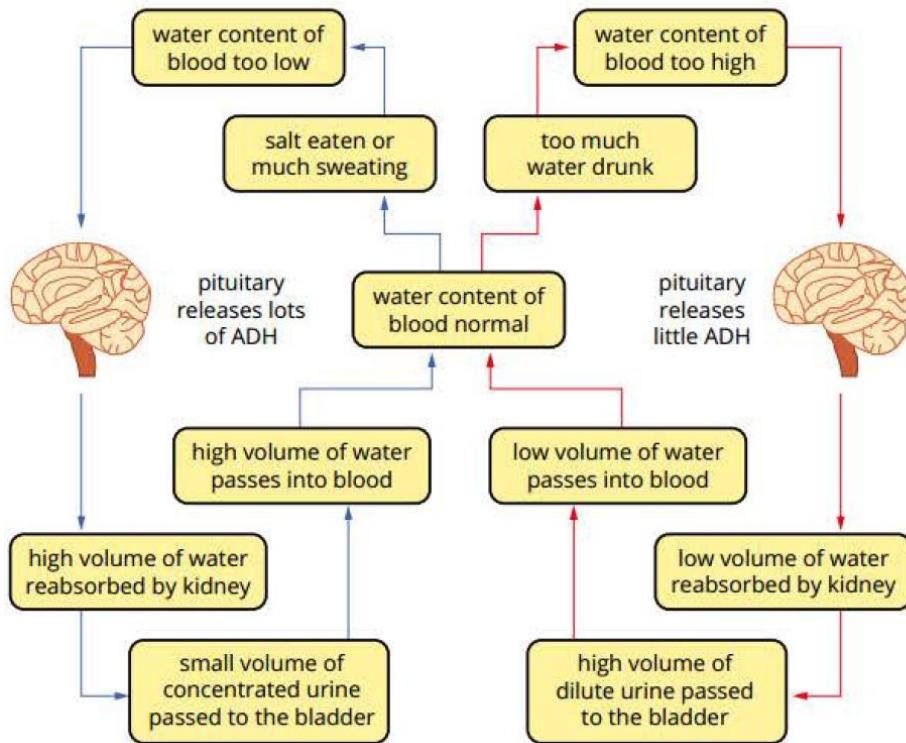
## Osmoregulation + Aldosterone

Aldosterone is a hormone responsible for regulating sodium and potassium to regulate blood volume.

Aldosterone is released from adrenal glands.

When low blood volume is detected, more aldosterone is released to increase sodium reabsorption into the blood and potassium excretion into the urine.

Increased sodium concentration increases blood volume and pressure.



# Plant Responses to Temperature Change + Water Maintenance

## Temperature Change

- **Radiation** - shiny leaves reduce the amount of heat being absorbed by plants (e.g. pigface)
- **Orientation of leaves** - vertical orientation (hanging) reduces surface area exposed to light ray and therefore heat (e.g. eucalyptus)

## Case Study: Eucalyptus Tree

### Behavioural Adaptation:

The eucalyptus tree releases seeds after there has been a fire. All the nutrients for the seeds to germinate are found in the ash bed, also known as the "ash bed effect". This gives the eucalyptus trees less competition from other plants who cannot survive after fire conditions meaning the young trees will have a better chance of survival.

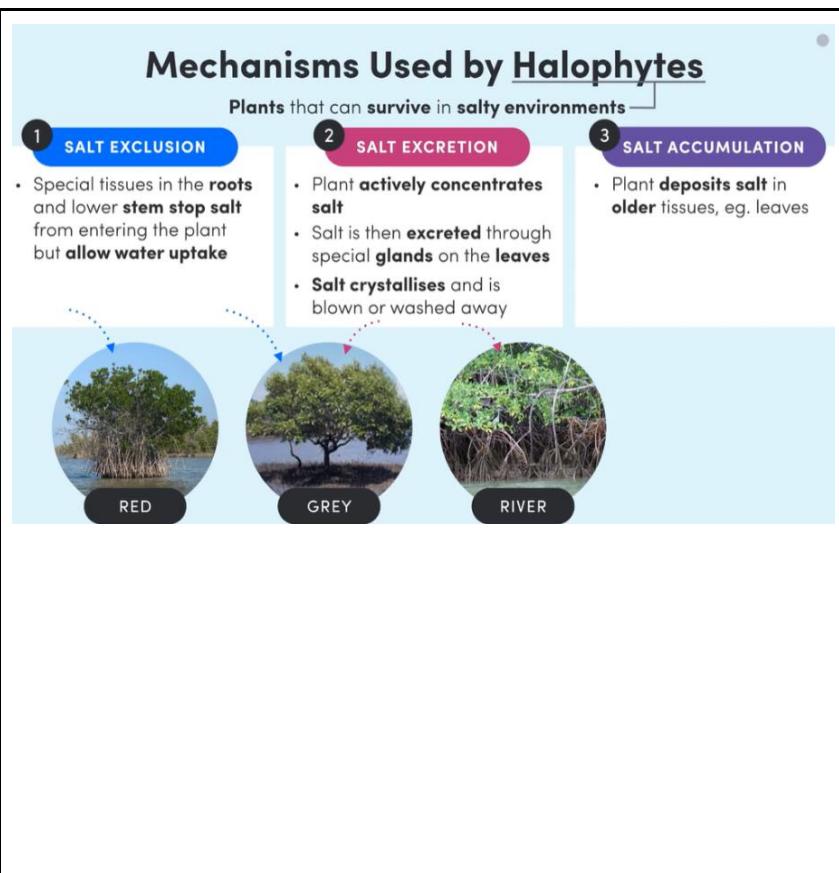
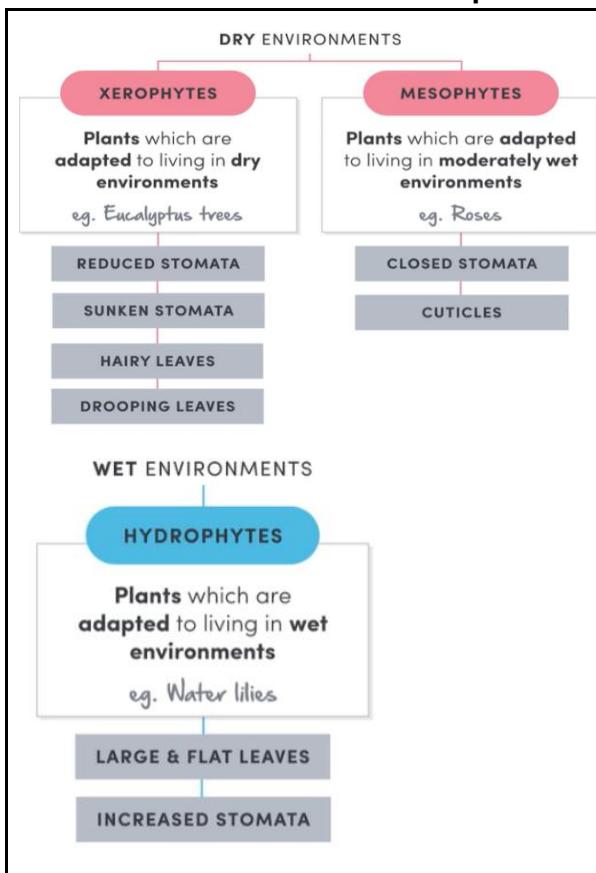
### Structural Adaptation:

The leaves of the eucalyptus tree hang vertically. By hanging vertically, it reduces the amount of light that the tree is exposed to, thus reducing transpiration.

### Physiological Adaptation:

The leaves of the eucalyptus tree contain toxic compounds. This is so only a few animals can eat the leaves of this tree. These animals are the koala and the greater glider.

## Water Maintenance + Osmosis in plants



# The Nervous System

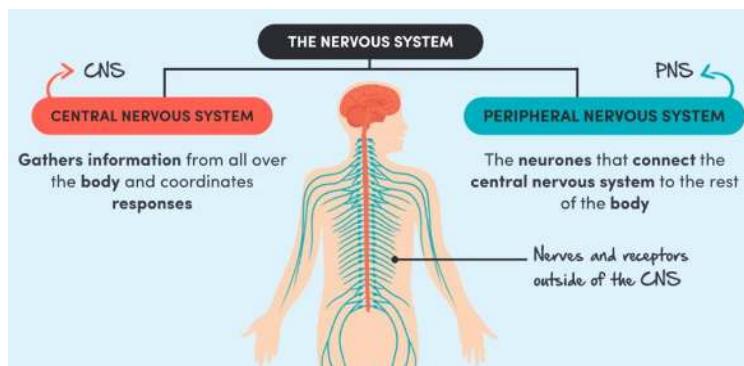
The nervous system works alongside the endocrine system to maintain homeostasis. The nervous system allows organisms to take information from the environment and responds by passing information around the body through a network of neural pathways.

## Overview of the Nervous System:

The nervous system takes messages from the receptors. It interprets the correct response to the information and finally sends messages to effectors in order to respond to stimulus.

It carries these messages as nerve impulses, along neurons. Nerve impulses travel between neurons to deliver information. Receptors and receptors are found all around the body, and there is an extensive network of neurons to carry messages to where they need to go.

The nervous system is the system that allows organisms to take information from the environment and responds by passing information around the body through a network of neural pathways.



## Components of the Nervous system:

### PNS

- **Somatic** : voluntary, conscious portion of the nervous system and is made up of nerves that connect to the skin, sensory organs and skeletal muscles
  - Function - processing sensory information that arrives via external stimuli (hearing, touch, sight). It is also responsible for controlling skeletal muscles.
- **Autonomic** : involuntary, unconscious portion of the nervous system and is made up of nerves that connect to the cardiac muscle in the heart and in the smooth muscles in the organs.
  - Function : controlling heart rate, digestion, salivation, sweating, pupil diameter, etc.

### Branching from Autonomic PNS

- **Parasympathetic (when you are calm and relaxed)**
  - Definition - the nervous system realises you're not in danger and releases acetylcholine (neurotransmitter).
    - 'The rest and digest system'
  - Effectors + Response - the heart slows to normal, the digestive system continues to digest food.
- **Sympathetic (when you are stressed)**
  - Definition - the nervous system realises you are in danger and releases adrenaline
    - 'Fight or flight response'
  - Effectors + Response - adrenaline tells the heart to speed up in order to get the blood pumping around your body so that your muscles have better access to oxygen. Digestion is stopped to focus energy towards survival

# Neurons

**Neurons** - functional units of the nervous system which carry signals throughout the body

**To remember the structure of a neuron, memorise CDAMS**

C - cell body

- Main spherical part of neuron
- Contains nucleus

D - dendrites

- Branch off cell body
- Carry information toward cell body

A - axon

- Long thread-like projection
- Carry information away from the cell body

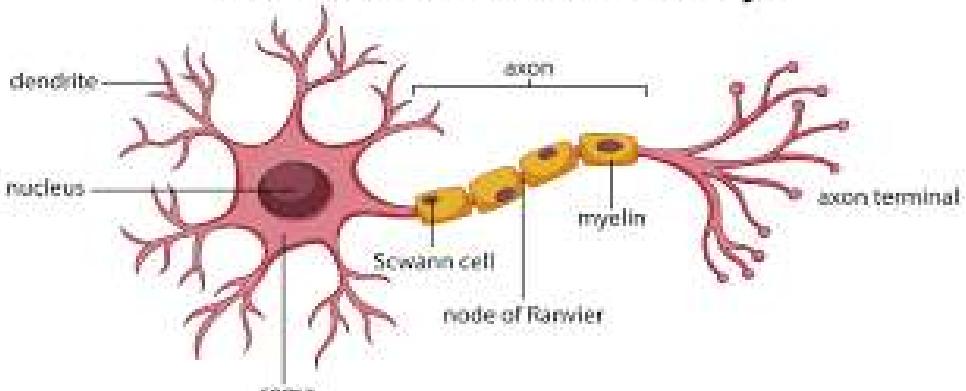
M - myelin sheath

- Coats axon to protect and insulate

S - synaptic knobs

- Branches from the end of the axon

## Neuron Anatomy



### SENSORY NEURONS



Transmit information from receptors to the CNS

### MOTOR NEURONS



Transmit information from the CNS to effectors

### INTERNEURONS



Transmit information between sensory and motor neurones within the CNS

# The Endocrine System

**Definition:** a group of glands that secrete hormones. The endocrine system is controlled via the stimulus response model.

**Hormones:** is a small chemical which causes a response in another region of the body. In other words, hormones are chemical messengers. Hormones are produced by glands.

The transportation of hormones occurs mainly in the circulatory system. Note that the lymphatic system also plays a role in hormone transportation.

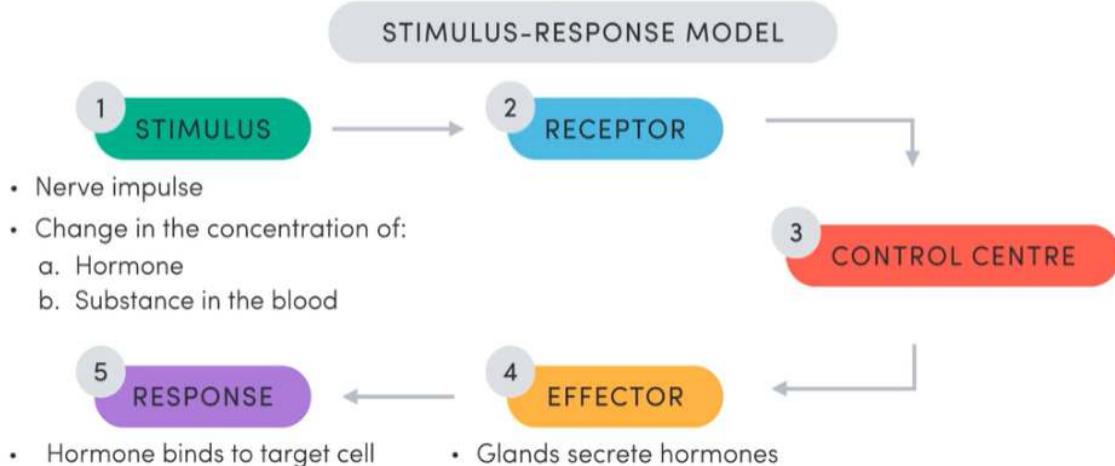
- Hormone diffuses out of glands and directly into the blood to be taken around the body by the circulatory system.
- Each hormone will only bind to specific receptors for that hormone found on target cells.

Compared to the Nervous System, the Endocrine System:

- Slower communication of message to effector
- Hormones can have effects on tissues that are widely distributed throughout the body
- A tissue just needs hormone receptors in order to be affected
  - Eg. Insulin - acts on liver, muscle and fat cells throughout the body to maintain glucose levels

## Summary

Groups of specialised cells      Made up of **glands** which secrete **hormones**      Chemical messengers



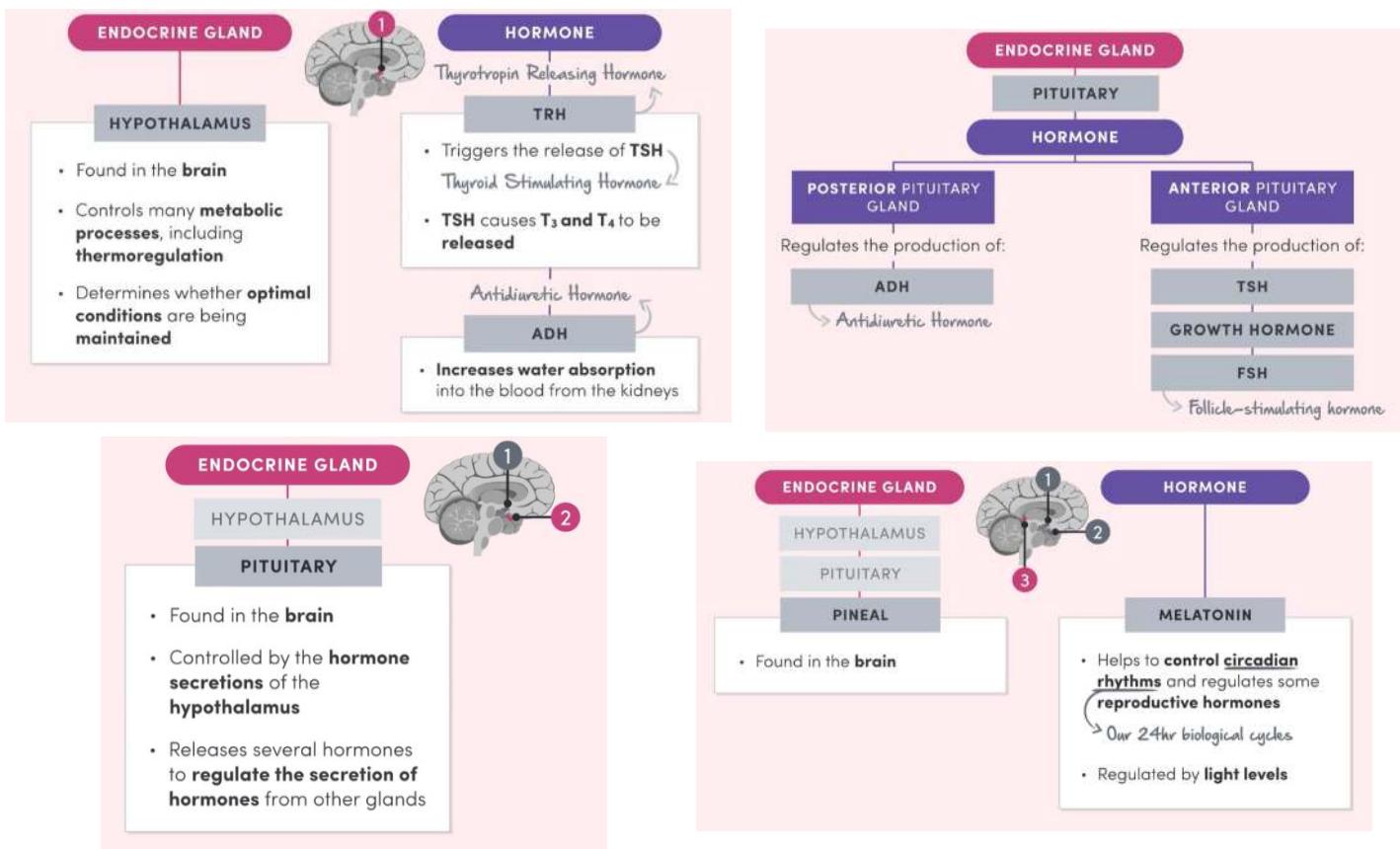
# Endocrine Glands + Hormones

## Why is it important to regulate internal temperatures?

The main reason most organisms are found within a certain temperature range in particular environments is due to the specificity of their enzymes. If the weather is too cold, the enzyme is stable but will not work while in very hot temperatures the enzyme becomes unstable and will denature.

### Hypothalamus:

The hypothalamus can detect internal change such as temperature change and release hormone by pituitary glands for homeostasis



## Summary

ENDOCRINE GLAND	HORMONE
	<ul style="list-style-type: none"> <li>Hypothalamus</li> <li>Pituitary</li> <li>Pineal</li> </ul>
	<ul style="list-style-type: none"> <li><math>T_3</math>, <math>T_4</math></li> <li>Parathyroid</li> </ul>
	<ul style="list-style-type: none"> <li>Thymosin</li> </ul>
	<ul style="list-style-type: none"> <li>Glucagon, Insulin</li> </ul>
	<ul style="list-style-type: none"> <li>Aldosterone, Adrenaline, Noradrenaline</li> </ul>
	<ul style="list-style-type: none"> <li>Progesterone, Estrogen, Testosterone</li> </ul>

## Pancreas gland:

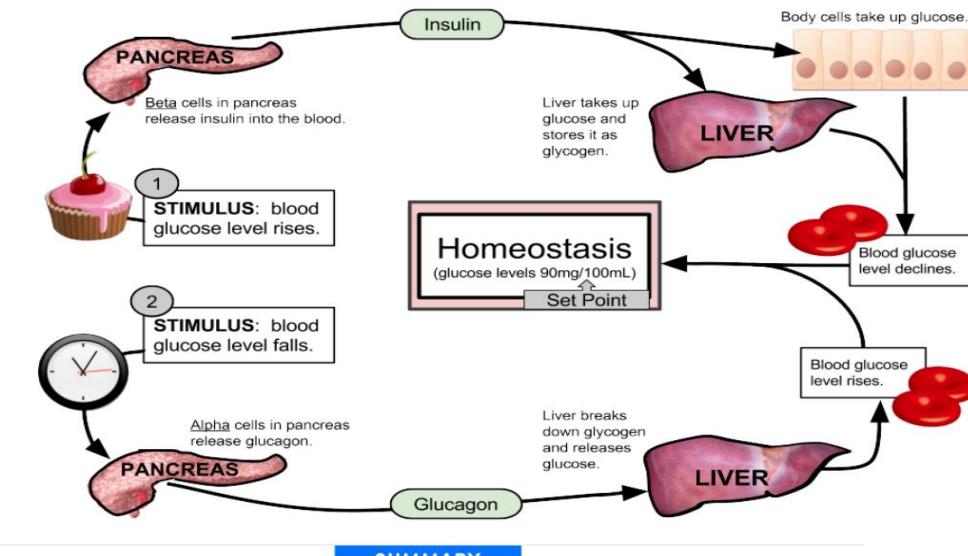
- Found behind stomach
- Controls blood glucose levels
- Glucose is the main source of energy for cellular respiration
  - (Recall: Glucose + Oxygen → Carbon dioxide + Water + ATP )
- Glucose level needs to be maintained within a certain range for the body to function properly.

## High blood glucose level is detected by Beta cells in pancreas

- Insulin will be released to uptake glucose into the liver if the glucose level is high.
- Hormone insulin is secreted to induce the uptake of glucose by the liver
- Glucose is then stored in the liver as glycogen
- The process is called Glycogenesis
- Failure to manage glucose may cause hyperglycemia in Diabetes

## Low blood glucose level is detected by Alpha cells in pancreas

- Hormone glucagon is then secreted to induce the release of glucose by the liver
- Glycogen in the liver is converted into glucose to be released
- This process is called Glycogenolysis
- Failure to manage glucose may cause hypoglycemia and fainting



→ released when the glucose is GONE!

	Glucagon 	Insulin 
<b>Effect</b>	Increase BGL	Decrease BGL
<b>Released by?</b>	Alpha cells (pancreas) 	Beta cells (pancreas) 
<b>Target cells</b>	Liver, muscle cells	Liver, muscle, fat cells
<b>Mechanisms</b>	<ul style="list-style-type: none"> <li>• Increase conversion of <b>glycogen to glucose</b> (liver, muscle cells)</li> </ul>	<ul style="list-style-type: none"> <li>• Increase conversion of <b>glucose to glycogen</b> (liver, muscle cells)</li> <li>• Increase uptake of <b>glucose</b> (liver, fat cells)</li> <li>• Increase conversion of <b>glucose to fatty acids</b> (liver cells)</li> </ul>

# Comparing the Nervous and Endocrine System

	Nervous System	Endocrine System
<b>Cells involved</b>	Neurones	Glands
<b>Signal</b>	Electrical (impulse)	Chemical (hormone)
<b>Signal carrier</b>	Neurones	Blood/lymph
<b>Speed of signal transmission</b>	Fast	Slow
<b>Size of the response generated by the signal</b>	Localised (a specific cell or tissue)	Systemic (cells or tissues throughout the body)
<b>Duration of response</b>	Temporary	Long-lasting

## Key Notes:

- The nervous system transmits electrical impulses along neurons.
- The endocrine system delivers hormones using the lymphatic and circulatory systems.
  - The nervous system creates faster and more localised responses compared to the endocrine system.
  - Responses created by the nervous system are short-lasting whereas the endocrine system creates permanent or long-lasting effects.
- The nervous and endocrine systems work independently or together to coordinate homeostatic processes, including:
  - Thermoregulation
  - Osmoregulation
  - Glucose homeostasis.

# Types of Non-Infectious Diseases

Non-infectious diseases are diseases that are **not caused by a pathogen** and **cannot be transmitted** between organisms.

Causes:

- Genetic disorders
- Environmental factors
- Cancers
- Nutritional disorders

## Genetic Disorders

Definition - altered or incorrect expression of a gene that causes disease. Due to this, the production of the protein coded for by that gene is altered.

The effect of genetic disorders vary depending on the nature of the mutation and which genes and chromosomes are affected.

Example - Cystic Fibrosis

- Caused by a mutation in the CFTR gene
  - caused the CFTR channel in the outer cell membrane to be faulty
  - causes sticky mucus to build up
  - leads to breathing problems and increased risk of infection

## Environmental Exposure

Definition - Environmental factors can trigger diseases within an organism's lifetime

Hypersensitivity Reaction - the overreaction of the immune system in response to antigens in the environment (eg. allergies)

Genetic Disorders - the risk of spontaneous mutations in genes is increased in response to mutagens (eg, radiation, chemicals and toxins)

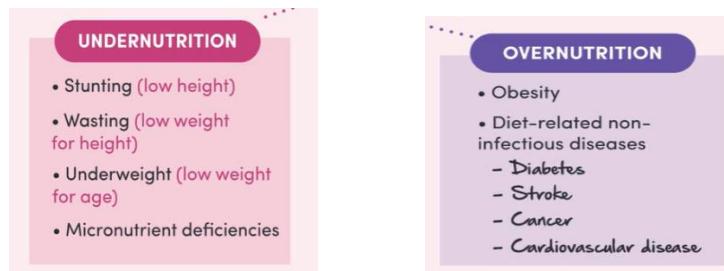
## Cancer

Definition - a group of diseases that involve unregulated and abnormal cell growth and division. Cancer can be caused by random mutations or environmental agents

**Carcinogen** is the term used to refer to cancer-causing agents. (tobacco smoke → lung cancer; UV radiation → skin cancer)

## Malnutrition

Definition - a deficiency, imbalance or excess of carbs, fats, proteins, vitamins, minerals and water



# Epidemiology

## What is it?

Definition - the study of the distribution, patterns and causes of disease in a population.

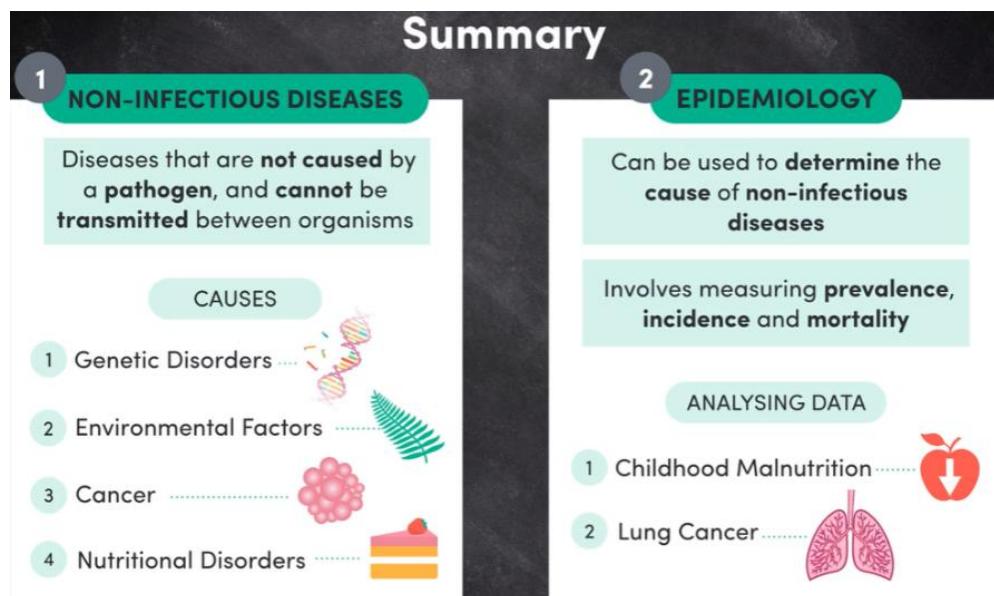
- Identify association between risk factors and disease
- Helps to make future predictions about disease distribution
- Helps to shape preventive measures

Epidemiology collects data on a disease with a high prevalence, incidence or mortality to try and determine what caused it

**Prevalence** - the number of living people diagnosed with a disease at a given time (the number of people with the disease)

**Incidence** - the number of new cases of a disease diagnosed within a specific time period (the rate at which new people are being infected)

**Mortality** - the number of deaths occurring as a result of the disease during a specific time period



# Epidemiological Studies

## Definition:

An epidemiological study involves the collection and statistical analysis of large quantities of data.

The data collected can be used to:

- Identify the cause of a disease or associated risk factors
- Determine patterns in the frequency or distribution of disease
- Compare changes over time
- Estimate trends
- Predict possible outbreaks in the future

Collecting data from lots of people helps to increase the chance that any association found between a disease and a particular factor is representative of a real relationship between the two.

## Types of Epidemiological Studies:

Analytical Studies - Analytical studies aim to test a specific hypothesis.

→ This is usually about the cause or risk factors associated with a disease.

→ Analytical studies can often show correlations between a risk factor and disease.

Descriptive Studies - Descriptive studies aim to determine patterns in the way a disease is distributed.

→ This can involve identifying where a disease occurs in the world or which groups of people are affected by the disease.

Intervention Studies - Intervention studies aim to measure the effectiveness and safety of a health intervention.

→ This may involve testing the effectiveness of a particular drug treatment, or recalling certain foods (depending on what is being studied)

## How data is collected in an Epidemiological Study:

- Interviews
- Questionnaires

The precise method of data collection comes down to the target group and aim of the study

→ For example, it would be inappropriate to conduct interviews over the phone if the target group are people with hearing impairments

collecting large data sets is crucial to ensure any associations between a disease and various factors are real

1. **Diagnostic stage:** where the presence of the disease is confirmed.
2. **Descriptive stage:** where the population at risk and distribution of the disease is described.
3. **Investigative stage:** where studies are implemented to investigate whether the evidence supports a hypothesis about the disease.
4. **Experimental stage:** where experiments are performed under controlled conditions to test the hypothesis.
5. **Analytical stage:** where the results are analysed.
6. **Decision-making stage:** where the knowledge gained in the previous phases is used to develop health programs or services, or direct further research.

# Evaluating Epidemiological Studies

## Limits on the study of epidemiology:

Epidemiological studies are great because they can identify:

- cause(s) of disease,
- emerging patterns and issues,
- and inequalities between different groups, which can be defined according to gender, age or economic status.

However, it's important to keep in mind that epidemiology simply studies that **pattern of health, not total health.**

Epidemiology fails to:

- **Provide a holistic view of health**, as it primarily focuses on physical health.
- **Explain sociocultural factors** that contribute to negative health behaviours.
- **Explain why health inequalities exist** between different groups.

→ For example, epidemiology shows that 17–25-year-old men have a higher risk of motor vehicle accidents than any other demographic. It also identifies lots of risk factors that are associated with these accidents, including speeding and alcohol.

However, epidemiology can't explain why young men are more likely to engage in these risky behaviours. This is where other fields of study (such as sociology and psychology) get involved. All this information can then be used to build public health campaigns that aim to reduce motor vehicle accidents in young men.

Overall, the field of epidemiology itself can't give us all of the answers for how to improve the health of populations. But keep in mind that everything starts with epidemiology, and in combination with other fields of study it plays a key role in improving health

## Benefits to Governments:

Epidemiology benefits governments by:

- Showcasing priority areas of concern.
- Informing methods of prevention and control.
- Directing public health campaigns.

Based on epidemiological information, we can determine priority areas of concern. This information is then used to inform the management of disease and to improve methods of prevention and control.

In particular, governments used epidemiological information to help construct public health campaigns.

Measuring how individuals are affected by disease also informs governments on how well the country's health care system is performing. Comparing changes over time, in particular, helps to estimate trends and predict future outbreaks.

Ultimately, this can help to maximise the use of resources and public health infrastructure so that health is improved as efficiently as possible

## Benefits to Individuals:

Epidemiology benefits individuals indirectly through the implementation of public health campaigns.

- Participating individuals are particularly benefited, as they may become more aware of their health or receive increased health updates that they otherwise wouldn't have.

# Conventional Cancer Treatments

## **Chemotherapy:**

Chemotherapy involves administering drugs which target and kill rapidly multiplying cells.

Cancer cells are known for their ability to grow really quickly, often forming tumours which is why it is essential to kill the cells quickly and effectively.

However, keep in mind that some normal, healthy cells (such as bone marrow and hair follicle cells) also grow quickly. As a result, these healthy cells are also destroyed by chemotherapy and this can have a terrible effect on a patient's health and wellbeing.

## **Radiotherapy:**

Radiotherapy involves using high doses of high energy radiation to kill cancer cells present in a tumour. The radiation achieves this by damaging the DNA inside of the cancer cells.

Remember, DNA is like a cell's instruction manual. So by damaging the DNA, radiation prevents the cell from being able to survive, grow or divide.

## **Surgery:**

Surgery can be used to physically remove a tumour from the body.

Although most of the tumour is removed, it can be very difficult to ensure that all cancer cells have been eliminated. On top of this, there are risks associated with any surgical procedure, such as infection and prolonged recovery time.

# Cancer Immunotherapy

## Defining Immunotherapy:

The immune system defends our body from pathogens, such as viruses and bacteria. On top of this, it also has a role in defending us from any of our own cells that might be harmful to our overall health, such as cancer cells. Immunotherapy is a type of treatment that harnesses the immune system in order to fight diseases such as cancer.

**Passive immunotherapy** involves the administration of agents that are otherwise a natural component of the immune system in order to enhance a patient's existing anti-cancer response.

→ Examples of these agents include antibodies and T lymphocytes.

**Active immunotherapy** attempts to stimulate the patient's own immune system to attack cancer cells.

→ Examples of active immunotherapy include cancer vaccines and targeting specific antigen receptors using cytokines.

→ An example is monoclonal antibodies (mAbs): antibodies produced by the same B lymphocyte. There are three types of mAbs to know about: standard, conjugated and bispecific.

Example: Cancer Vaccines

- Cancer vaccines contain antigens from cancer cells or cancer-causing viruses.
- Cancer vaccines work by stimulating a person's own immune system to produce immunological memory (think T and B memory cells) against cancer cells or cancer-causing viruses.
- Cancer vaccines can be categorised based on their purpose; so based on whether they are preventative or therapeutic.
  - Preventative cancer vaccines aim to stop cancer from developing in the first place.
  - Therapeutic cancer vaccines are made up of antigens for a particular type of cancer cell. These are given to people who already have cancer, in order to boost their immune response to the cancer cells.

## Future Directions for Cancer Immunotherapy:

Cancer immunotherapy has been a revolutionary cancer treatment. In clinical trials, it has been shown to be capable of permanently eliminating cancer with minimal side effects.

- Combinatorial Immunotherapy
  - There have been studies which have suggested that combining different immunotherapies can improve treatment. But, there are still questions to be answered here.
    - For example, there is still uncertainty in terms of how much of each immunotherapy should be administered (dosage) and how to identify the best combinations for the treatment of a particular patient or cancer type.
  - On top of this, the potential for immunotherapy combined with other types of treatment (such as chemotherapy or radiotherapy) hasn't really been looked into at all
- Personalised Immunotherapy
  - Personalised cancer vaccines are one of the pioneering examples.
  - They're a type of therapeutic cancer vaccine, used to treat people who already have cancer.
  - They contain a modified version of the patient's own cancer cells and they strengthen the immune response by making the cancerous cells more obvious to the immune system.

# The Ear

## Outer Ear:

- **Pinna** - fleshy, external tissue which consists of a flap cartilage and skin
  - Function - collect and funnel sound waves into the ear canal
- **Ear canal** - a passage comprised of bone and skin
  - Function - leads sound waves to the eardrum
- **Tympanic Membrane (Ear Drum)** - thin membrane between the outer and middle ear
  - Function - vibrates in response to sound. Specifically, it vibrates at the same frequency as the sound wave.  
Ear drums transfer sound vibrations to the ossicles.

## Middle Ear:

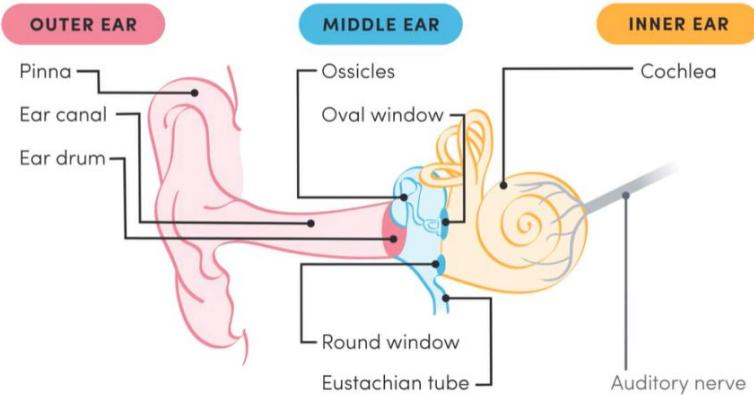
- **Ossicles** - structure of 3 bones (Malleus, incus, stapes AKA hammer, anvil, stirrup)
  - Function - amplify and transmit vibrations from the eardrum to the oval window
- **Oval window** - thin, flexible membrane
  - Function - transmits vibrations from the stapes to the fluid in the cochlea
- **Round window** - thin membrane between middle and inner ear
  - Function - bulge outwards to allow equalisation of pressure in the cochlea when the oval window vibrates
- **Eustachian Tube** - connects the middle ear to the pharynx at the back of the throat
  - Function - equalises air pressure on both sides of the eardrum so that it is not overly stretched

## Inner Ear:

- **Cochlea** - snail-shaped, spiral tube filled with fluid.
- **Organ of Corti** - The middle chamber of the cochlea consists of hair cells attached to the basilar membrane.
  - Function - hair cells act as sound receptors.  
→ When bent, they generate nerve impulses
- The vestibular apparatus is involved in maintaining balance
- **Auditory nerve** - transmits nerve impulses to the brain for interpretation

## \* How do these hair cells get bent?

When the oval window vibrates, it causes the fluid in the cochlea to vibrate, similar to waves. The basilar membrane flexes in response to this fluid movement, bending the hair cells as they rub against this membrane. Through this, the hair cells generate nerve impulses which are then transmitted to the brain via the auditory nerve so that the sound can be processed and interpreted.



# Hearing Loss

## Conductive Hearing Loss:

Treatment

Typically corrected with medication or surgery

### DEFINITION

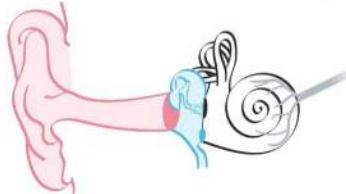
Deafness which is caused by **damage** to the **outer or middle ear**

### RESULT

Results in **reduced or muffled sound**

### CAUSES

- Malformed structure at birth
- Aging
- Noise strain
- Injury
- Infection
- Blockage due to earwax, a tumour or foreign item



## Auditory Processing Disorder:

There are limited therapies available that can treat auditory processing disorders, although educational intervention can help

### DEFINITION

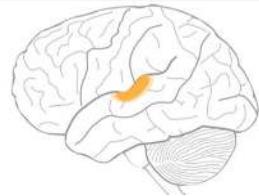
Not due to **damage** or defects in the **structure** of the **ear**

### RESULT

Inability to detect the **direction** of a **sound**

### CAUSES

- Processing problems in the **auditory areas** of the brain



## Sensorineural Hearing Loss:

Treatment

Cannot be corrected medically or surgically

Damage to the hair cells are permanent

### DEFINITION

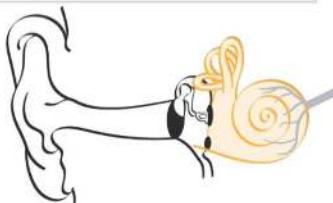
Deafness which is caused by **damage** to the **inner ear/nerve pathways** from the **inner ear** to the **brain**

### RESULT

Results in **faint or muffled sound**

### CAUSES

- Malformed inner ear at birth
- Exposure to damaging noise can cause the hair cells in the cochlea to become fatigued/kill them
- Head injury



## Tinnitus:

Tinnitus is a broad term for hearing a ringing in the ears

### DEFINITION

When a **sound** is heard that **isn't there**

### RESULT

Can include hearing a **ringing** in the ears: **buzzing, hissing**

### CAUSES

- Exposure to loud sound
- Injury
- Aging



# Treatments for Hearing Loss

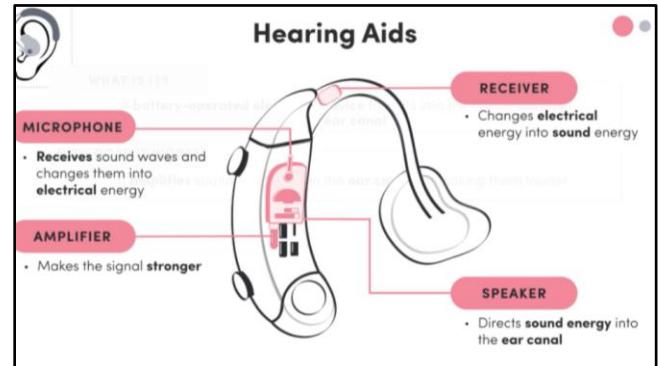
## Hearing Aids:

### What is it?

- A battery-operated electronic device that fits into the hollow outside of the ear canal

### How does it work?

- It amplifies sound waves within the ear canal
- The eardrum, ossicles and cochlea must still have an adequate level of function so that the sound can be registered and converted into an electrochemical signal
- Can be used for people with conductive or sensorineural hearing loss



Advantages	Disadvantages
<ul style="list-style-type: none"><li>• Relatively cheap</li><li>• Easy to install - since no surgery is required</li></ul>	<ul style="list-style-type: none"><li>• They amplify all sounds in an environment (including background noise which can irritate and cause pain)</li><li>• It does not help people with severe damage to the inner ear. Auditory nerve</li></ul>

## Bone Conduction Implants:

- Works by creating sound vibrations, that are conducted directly to the cochlea
- Used for treating conductive hearing loss
- This device can bypass a dysfunctional outer and middle ear but the inner ear must be working

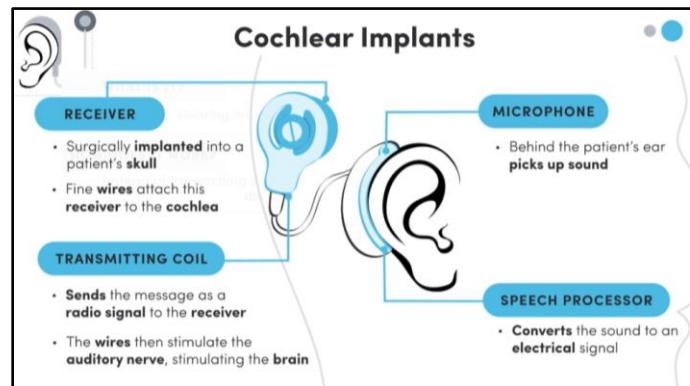
## Cochlear Implants:

### What is it?

- Used by people with profound hearing loss  
→ profound damage to any region of the ear

### How does it work?

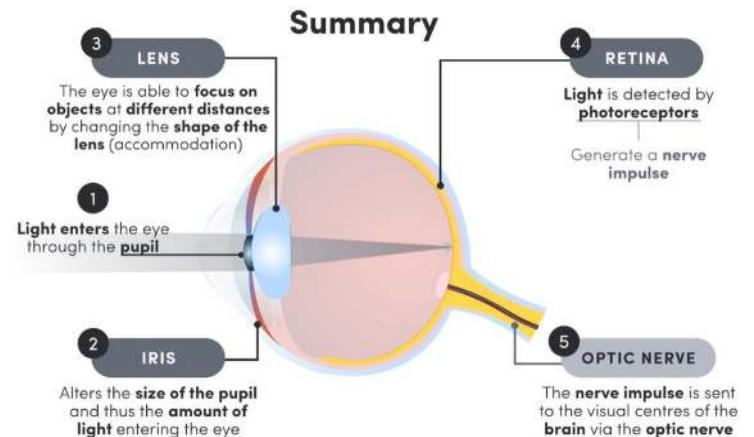
- Instead of transmitting sound waves, it converts sound into electrical signals and directly stimulates the auditory nerve



# The Eye

## Overview of the eye:

- Visible light is a kind of electromagnetic radiation
  - Energy which travels through the universe as a wave
  - Light waves can have different amounts of energy
  - We perceive these differences in energy as different colours
- When light meets an object, it's either:
  - Absorbed
  - Reflected



## Key Parts of the Eye:

**Iris** - Helps deal with light at different intensities

- Structure - ring of pigmented muscle tissue
- Function - Works to control pupil size and hence the amount of light entering the eye.
  - Bright light : can damage the photoreceptors
  - Dim light : the photoreceptors are not being stimulated and an image cannot be formed

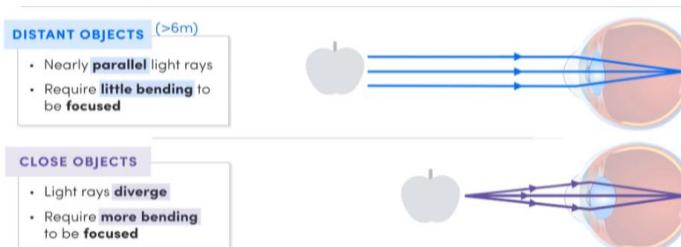
**Lens** - Helps focus on objects at different distances away from us

- Structure - transparent, biconvex protein disk. Has a bulging shape and is made out of protein
- Function - adjusting its thickness to bend light so that it focuses directly onto the retina

**Accommodation** - the process in which the lens changes shape to maintain a clear focus on objects at varying distances

**Retina** - responsible for detecting light and converting the information into nerve impulses

- Structure - thin layer of photoreceptor cells, which lines the inner back surface of the eye
  - Photoreceptor cells are specialised neurons that contain light-sensitive pigments (photoproteins)
  - RODS: Does not detect colour and works best at low light levels (night)
  - CONES: Detects colour and works best in bright light (daytime)
- Function
  - After light has passed through the pupil, lens and jelly-like fluid, it hits the photoreceptor cells on the retina
  - When photoreceptor cells are exposed to light, their photopigments change shape
  - Photoreceptor cells generate an electrochemical signal that is sent to the brain to be interpreted



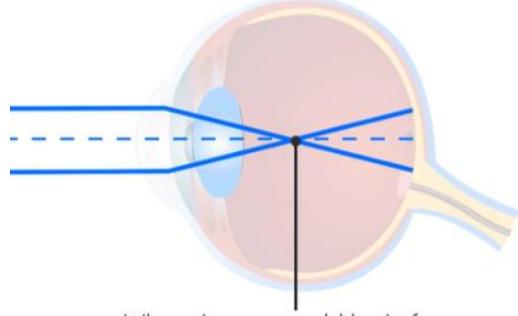
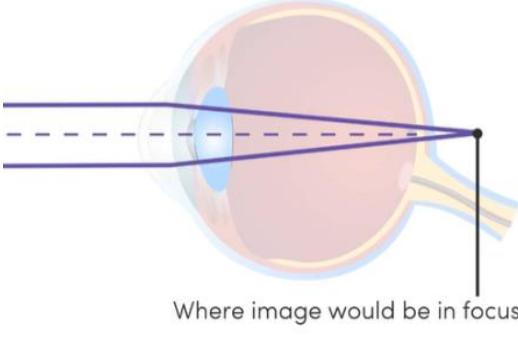
# Visual Disorders

## Refractive Errors

Refractive errors prevent light from falling on the retina correctly.

This causes blurry/ foggy vision, caused by misshapen lens or eyeball

Causes - thickness of the lens or the shape of the eyeball

MYOPIA - short sightedness	HYPEROPIA - long sightedness
<ul style="list-style-type: none"><li>• <u>Close objects are clear</u>, but distant objects are blurry</li><li>• The image is being focused <u>in front</u> of the retina</li></ul> <p>Causes:</p> <ul style="list-style-type: none"><li>• The lens is too thick = bends light too much</li><li>• The eyeball is too long = the retina is further away</li></ul>  <p>Where image would be in focus</p>	<ul style="list-style-type: none"><li>• <u>Distant objects are clear</u>, but close objects are blurry</li><li>• The image is being focused <u>behind</u> the retina</li></ul> <p>Causes:</p> <ul style="list-style-type: none"><li>• The lens is too thin = doesn't bend light enough</li><li>• Eyeball is too short</li></ul>  <p>Where image would be in focus</p>

## Retina Conditions

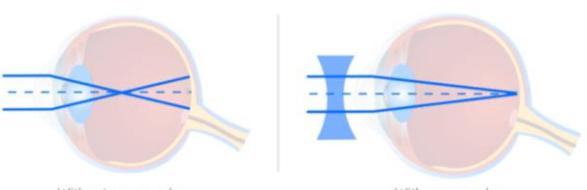
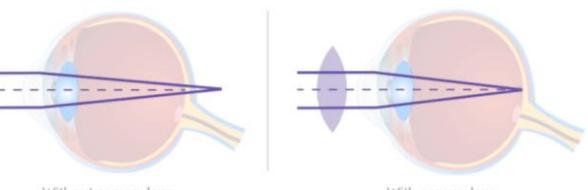
Damage to the retina causes loss of vision

Retinopathy	Macular Degeneration	Colour Blindness
<ul style="list-style-type: none"><li>• Damage to the retina</li></ul> <p>Causes:</p> <ul style="list-style-type: none"><li>• Changes to the blood vessels surrounding the area</li></ul>	<ul style="list-style-type: none"><li>• Caused by a distortion or absence of the fovea (central, most sensitive part of the retina)</li></ul>	<ul style="list-style-type: none"><li>• Occurs when one of the three types of colour detecting cone cells is absent or does not function properly</li><li>• Typically inherited</li><li>• Affects both eyes</li></ul>

# Treatments for Visual Disorders

## Glasses

- Glasses have lenses which are shaped in order to correct individual needs, by compensating for misshapen eye lenses or eyeballs
- Glasses cause light rays to bend before entering the eye so that the image focuses on the retina
- Two types of glasses: Concave (curved inwards) + Convex (bulging outwards)
- Contact lenses work the same way as glasses but are put directly on the eyeball

CONCAVE	CONVEX
<ul style="list-style-type: none"> <li>• Concave glasses correct MYOPIA</li> <li>• Causes light rays to <b>diverge</b> so that the image forms further back, falling on the retina</li> </ul>  <p>Without concave lens      With concave lens</p>	<ul style="list-style-type: none"> <li>• Convex glasses correct HYPEROPIA</li> <li>• Causes light rays to <b>converge</b> so that the image forms further forward on the retina</li> </ul>  <p>Without convex lens      With convex lens</p>

Glasses	Contact Lenses
<b>Pros:</b> <ul style="list-style-type: none"> <li>- Cheap</li> <li>- Easy solution to vision impairment</li> </ul> <b>Cons:</b> <ul style="list-style-type: none"> <li>- Inconvenient</li> <li>- Maintenance</li> <li>- Fragile and temporary</li> </ul>	<b>Pros:</b> <ul style="list-style-type: none"> <li>- Convenient</li> </ul> <b>Cons:</b> <ul style="list-style-type: none"> <li>- Expensive</li> <li>- Higher maintenance</li> <li>- Chance of infection</li> <li>- Difficult to insert and remove from the eyeball</li> </ul>

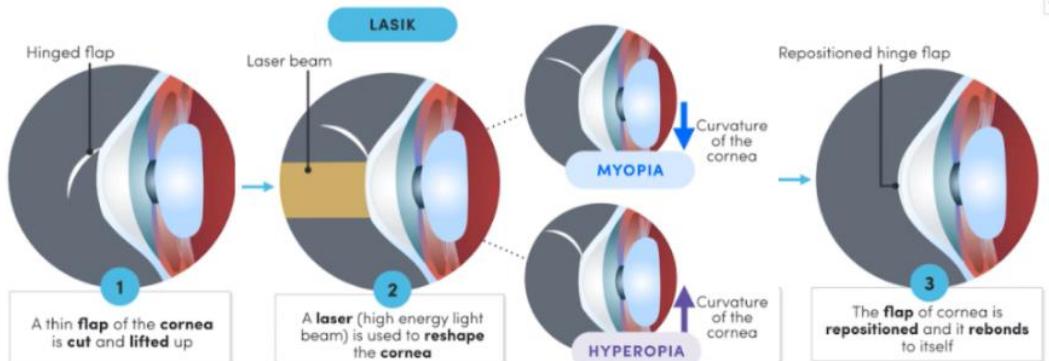
## Laser Surgery

### Pros:

- Long term solution
- Highly Effective
- Fast

### Cons:

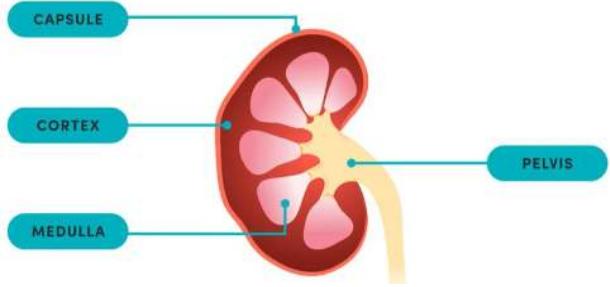
- Expensive, eligibility requirement
- Surgical risk
- Recovery time
- Permanent damage



# The Kidney

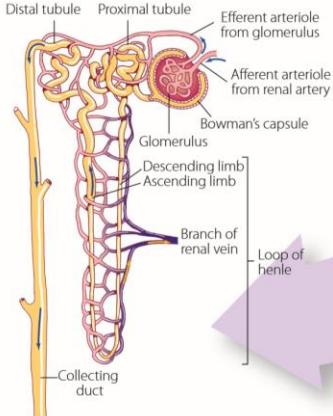
## Structure:

- Responsible for filtering the blood
- The kidneys excrete excess wastes, salt and water in order to maintain homeostasis



**Nephrons:** functional unit of the kidney.

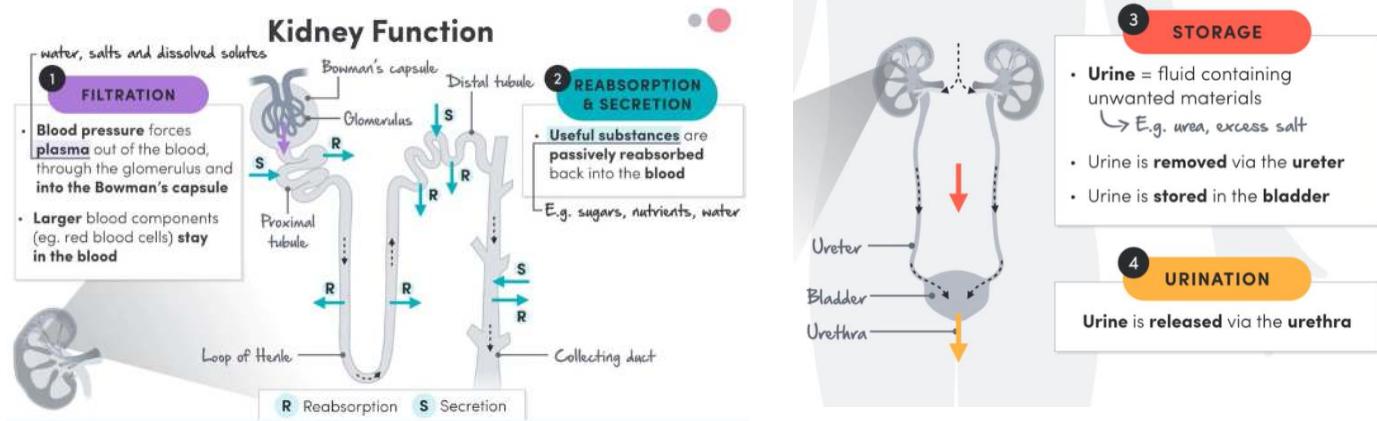
The structure that actually filters blood



Component	Function
Bowman's capsule	A cuplike sac that encloses a glomerulus; it functions to carry out the first step in filtration of blood from the urine.
Glomerulus	A ball of capillaries within the Bowman's capsule that acts as an 'ultra' filter
Long twisted tubules and Loop of Henle	The twisted tubules occur in the cortex; the Loop of Henle is the u-shaped part of the nephron and it extends from the cortex down into the medulla and back; the tubules play a role in the reabsorption of substances.
Collecting tubule	Long straight tubule which opens into the pelvis of the kidney; the wall of the collecting tubule may be permeable or impermeable to water. Permeability of the wall is controlled by ADH.

## Function:

- Blood enters the kidney via a blood vessel called the **renal artery**.
- Once in the kidney, the blood flows through a series of smaller and smaller arteries until it reaches the **glomerulus in the nephron**.
- Firstly, blood pressure forces plasma out of the blood, through the cells in the wall of the glomerulus and into the **bowman's capsule**. The larger blood components stay in the blood.
- Basically, substances are filtered out of the blood and into the nephron at the bowman's capsule.
- The fluid that passes into the capsule is referred to as the 'filtrate'. The filtrate then flows into the tubule. The tubule is semi-permeable; which allows any 'useful' substances - such as **sugars, nutrients and water** to pass back into the blood.
- By the end of the filtration process at the end of the tubule, is a liquid containing all of the unwanted materials that we call **urine**.



# Kidney Disorders

Kidney disorders refer to when the kidneys are damaged and do not function as they should. Disorders and diseases can vary from temporary disorders or to renal failure.

## Chronic Kidney Disorders

Glomerulonephritis	Diabetic Nephropathy	Polycystic Kidney Disease	Kidney Stones
<ul style="list-style-type: none"><li>- inflammation of the glomerulus</li><li>- occurs after a bacterial infection</li><li>- common type of kidney disorder</li></ul>	<ul style="list-style-type: none"><li>- Kidney damage caused by diabetes</li><li>- less efficient removal of wastes</li><li>- Larger substances can be filtered out the blood when they shouldn't be able to</li></ul>	<ul style="list-style-type: none"><li>- causes cysts to grow in the kidneys</li><li>- inherited condition</li><li>- cysts can enlarge and damage the kidneys</li></ul>	<ul style="list-style-type: none"><li>- forms when waste products crystallise into lumps</li><li>- can be painful and cause blockages</li></ul> <p>Risk factors:</p> <ul style="list-style-type: none"><li>→ dehydration</li><li>→ poor diet</li><li>→ hereditary factors</li></ul>

Effect:

- Prevent the kidney from effectively filtering the blood
- Metabolic wastes build up and salt and water levels become imbalance

## Treatments for Kidney Disorders

### Dialysis

The process of removing waste products and excess fluid from blood by an external machine called a dialyser.

#### How it works:

Blood is fed into the dialyser from an artery. Inside the dialyser, this blood flows through tubing made of a selectively permeable membrane that is immersed in dialysing solution. The features of both are super important to how the dialyser can work.

Semi permeable refers to the membrane that allows small molecules to move out of the blood. At the same time, it prevents any large molecules such as protein from leaving the blood.

The solution contains lots of minerals that promote the diffusion of appropriate substances into and out of the blood. It has a higher concentration of glucose and ions to limit their diffusion from the blood. At the same time, it has a low concentration of wastes to promote their diffusion out of the blood.

To maintain a steep concentration gradient, the dialysing solution is continuously replenished with fresh solution while the used solution is removed. The filtered blood is then circulated back into the body through a vein.

