Module 5: Heredity

5.1: Reproduction

How does reproduction ensure the continuity of species?

Reproduction ensures the continuity of species by ensuring the production of fertile offspring with genetic diversity for adaptability, through inheritance of genetic material.

Organism → Survives → Growth → Reproduction → Inheritance of genes → Continuity

[5.1.1] Mechanisms of Reproduction

ASEXUAL AND SEXUAL REPRODUCTION

Reproduction: The process of creating offspring, either sexually or asexually.

- ➔ **Sexual reproduction:** Reproduction which involves the combination of gametes, so that the offspring are genetically different to the parents.

- ➔ **Asexual reproduction:** Reproduction which does not involve the combination of gametes, so the offspring are identical to the parent.

	Advantages	**Disadvantages**
Sexual reproduction	✓ Variations in the population lead to continuity of species ✓ There are more variations, so species are better able to adapt to changing selection pressures ✓ It is less likely that a selection pressure will remove all of the species	× Requires more time and energy to carry out × Requires a mating partner, can not be done individually × Fewer offspring produced
Asexual reproduction	✓ Allows a species to rapidly populate an environment ✓ No requirement of mates ✓ Quick and not energy intensive, so it can be carried out when the parent organism is under stress ✓ There is no requirement to care for the offspring	× Lack of diversity between species of a population. Variations, and therefore adaptations, are limited × Reduces ability to adapt to selection pressures × May lead to extinction of the population, as one selection pressure can wipe out all species, as there is no variation between them

INTERNAL AND EXTERNAL FERTILISATION

Fertilisation: The fusion of gametes in sexual reproduction.

➔ **Internal fertilisation:** Fertilisation where the combination of gametes occurs within the female body.

➔ **External fertilisation:**Fertilisation where the combination of gametes occurs in the external environment.

INTERNAL FERTILISATION	✓ Increased likelihood of fertilisation as the gametes are in close proximity ✓ Less gametes produced, which reduces energy expenditure ✓ Offspring have a greater chance of survival as they are protected from the environment	× Fewer offspring are produced over more time × Mating rituals are time consuming and postpone copulation × Higher risk of sexually transmitted diseases/ infections × Fertilisation, gestation and parental care demand energy from the parent × Breeding has to be paused for gestation (pregnancy) period
EXTERNAL FERTILISATION	✓A greater number of offspring can be produced ✓ The behavioural process is simpler ✓ Prevention of dehydration as it occurs in an aquatic environment ✓ Reproduction can continue without pause for gestation ✓ Offspring are dispersed, meaning less competition ✓ Many gametes released, increasing chance of fertilisation ✓ No energy spent on parental care	× A large number of gametes must be produced. This requires a lot of energy × Can only occur in aquatic environments × The gametes are more susceptible to the risks of the open environment × External environmental conditions are unconstant, which may impact fertilisation × Absence of parental care can increase risk of predation and disease to offspring.

Comparison of internal and external fertilisation

Characteristic	Internal	Both	External
Gametes Released	Large number of sperm, Small number of eggs		Large number of both gametes
Fertilisation	Occurs in female reproductive system	Sperm fertilises egg	Occurs in external aquatic environment
Synchronisation of gamete release	Sperm is continuously produced, Eggs are produced over a cycle		Male and female gametes released simultaneously
Chance of fertilisation	High	Chance increases with proximity of gametes	Low
Result of fertilisation	Zygote (internal development)	The development of a zygote requires a watery medium	Zygote (external development)
No. of offspring	Small (but survival rate is high)	Both determined by: - No. of gametes produced - Survival rate	High (but survival rate is low)
Breeding frequency	Low, seasonal	Breeding frequency relies on environmental conditions	High
Parental care	Yes - Care of zygote during development - Care of offspring after birth		None

DEVELOPMENT

Development: The growth of the zygote.

When fertilised internally, development can be:

- ➔ **Oviparous:** External within an egg.
- ➔ **Viviparous:** Internal within the mother.
- ➔ **Ovoviviparous:** Internal within an egg within the mother.

Unisexual reproductive system Separate male and female individuals with separate reproductive systems.	✓ Less energy required to maintain 1 set of reproductive organs	× Mating rituals required, which take time
Bisexual/ hermaphrodite reproductive system Organisms have both male and female reproductive organs.	✓ Organism doesn't need a partner of the opposite sex to reproduce ✓ Beneficial for organisms that are sedentary	× A larger amount of energy is required to maintain 2 sets of reproductive organs

SEXUAL REPRODUCTION

IN ANIMALS

Gametes are produced by the parents by meiosis

↓

Each gamete is haploid (½ the total number of chromosomes)

↓

The sperm (male gamete) fertilises the oven/ egg (female gamete)

↓

The fusion of gametes results in a diploid zygote (has a complete set of chromosomes). The zygote has a unique combination of genetic material from both parents.

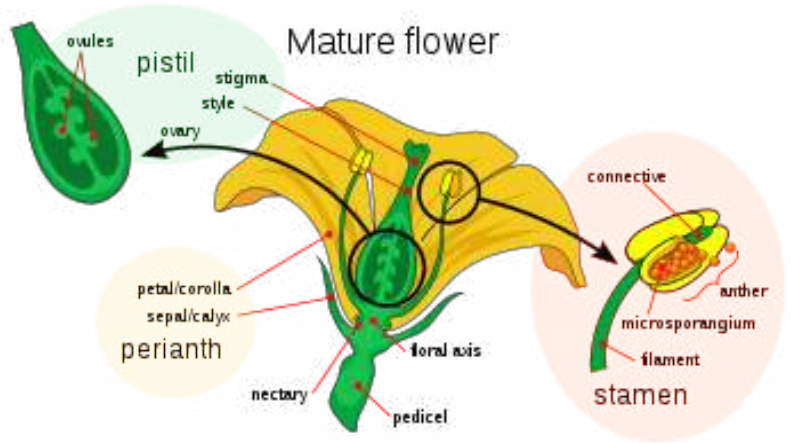
Animal fertilisation can be internal or external.

IN PLANTS

Plants are bisexual. In angiosperms, the:

MALE organs → Stamen (filament + anther)

FEMALE organs → Pistil (stigma + style + ovary)



Pollination

Pollen (the male gamete) is produced in the stamen, and held up at the anther

↓

The pollen is removed from the anther by the wind or by insects. It is transferred to the pistil of the same flower(self-pollination) or another flower (cross-pollination)

↳ [From the same plant or another one].

Fertilisation

The pollen falls on the stigma, and a pollen tube grows through the stile to the ovary

↓

The pollen migrates through pollen tubes, to the ovules

↓

The pollen fertilises the ovules

Development

The fertilised ovule develops into a seed, containing the embryo

↓

The seed protects the embryo

↓

The fruit around it provides it with nutrients

↓

Germination

When the fruit is removed, the seeds within it grow separate from the parent plant(s)

↓

The embryo lies dormant

↓

If on suitable soil, germination begins

↓

The embryo puts out its:

➔ Radical (first root) for absorption of water and nutrients

➔ Plumule (young stem) which develops leaves for photosynthesis

↓

The seed germinates into a seedling

Methods of pollination

METHOD	ADAPTATIONS FOR METHOD
● Wind	- Protruding stigma to catch more pollen - Light-weight, small pollen - Large pollen production - Flower is not colourful to conserve energy, as it doesn't need to attract pollinators
● Animals ○ Insects ○ Birds	- Colourful petals to attract pollinators - UV markings which insects are attracted to - Scent for attraction - Flower produces nectar to attract pollinators - Stigma and anthers within flower, energy not wasted in growing tall - Pollen is sticky, to attach to the pollinator and be dropped elsewhere

Methods of seed dispersal

METHOD	ADAPTATIONS FOR METHOD
• Wind	- Seeds are very light-weight
• Animals ○ Insects ○ Birds	- The seeds are *fruit bearing*, meaning that it is eaten by animals - Once the fruit is eaten, the seed is either - Left to germinate - Passes through the digestive system, excreted and germinates
• Self-dispersal	- The plant has an explosive mechanism, which propels the seed away from the mother plant - Propelling the seed to a different area reduces the competition for resources in one area, giving the offspring a greater chance of survival

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IN PROTISTS

Haploid (*n*) protists

Two haploid protist cells combine, and genetic material is combined in a new, fused nucleus. This produces a diploid zygote

↓

The zygote undergoes meiosis to produce 4 haploid daughter cells

Diploid (*2n*) protists

Adult cells undergo meiosis, producing 4 haploid gametes

↓

Gametes fuse during fertilisation to from a diploid zygote

↓

The diploid zygote will grow to maturity

IN FUNGI

Mitosis splits a diploid cell into haploid two daughter cells, which each contain a single set of chromosomes

↓

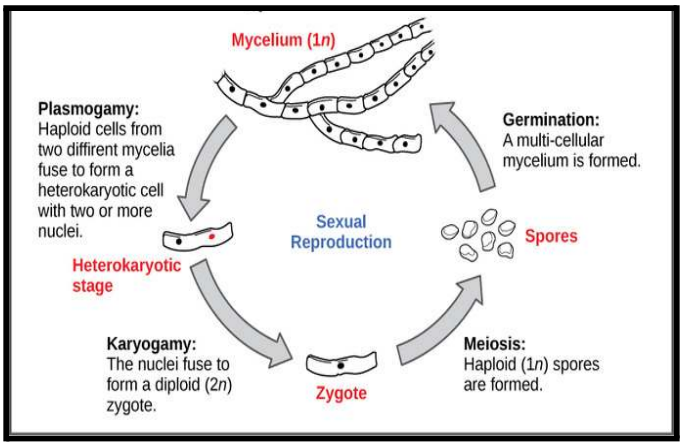
PLASMOLOGY: Two genetically different cells fuse together, combining their contents. This brings together two compatible haploid nuclei

↓

KARYOGAMY: The two haploid nuclei fuse into one diploid nucleus. This forms a zygote cell

↓

MEIOSIS: The zygote undergoes meiosis which produces haploid spores. These spores are distributed into the environment



ASEXUAL REPRODUCTION
IN PLANTS

Vegetative propagation

Method of asexual reproduction in plants, done by **vegetative organs**, which are modified parts of the plant.

Perennating organs: Underground organs which store food.

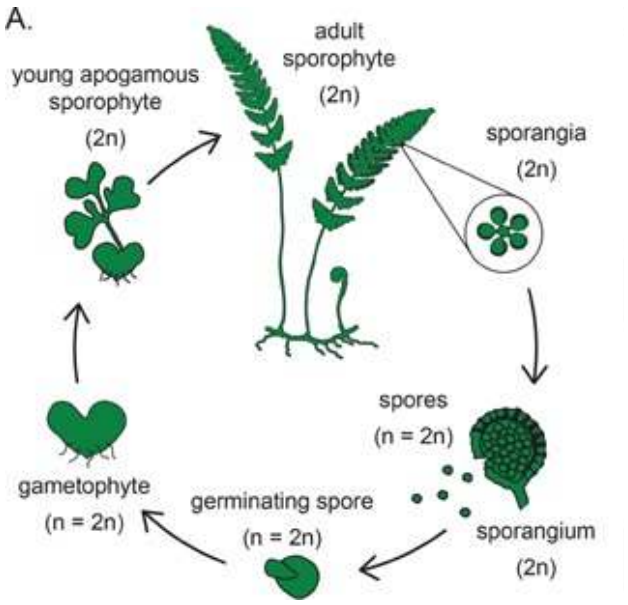
Modified part	Vegetative organ	Perrenating?
Stem	- Runners Underground stems that connect plants.	NO
	- Rhizomes Horizontal stems that store nutrients.	YES
	- Tubers Develop from swollen regions of a stem, or from buds/ eyes.	NO
	- Bulbs Scales of bulb store food. New plants can grow from the bulb.	YES
Roots	- Suckers Vertical growths that can resprout from underground.	NO

Apomixis (process)

Generative tissue gives rise to plantlets asexually.

Generative tissue includes:

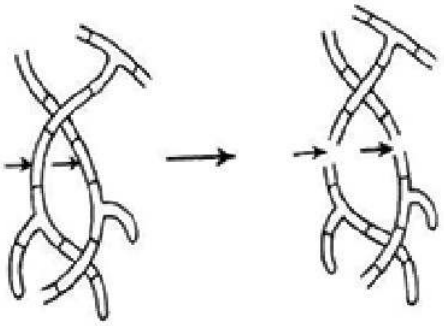
- Unfertilised ovules
- Specialised leaf tissue.



IN FUNGI

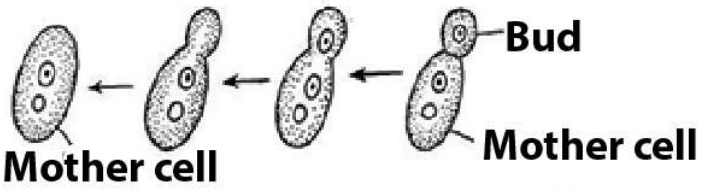
Fragmentation

Pieces of the fungal colony can break off and become separate colonies.



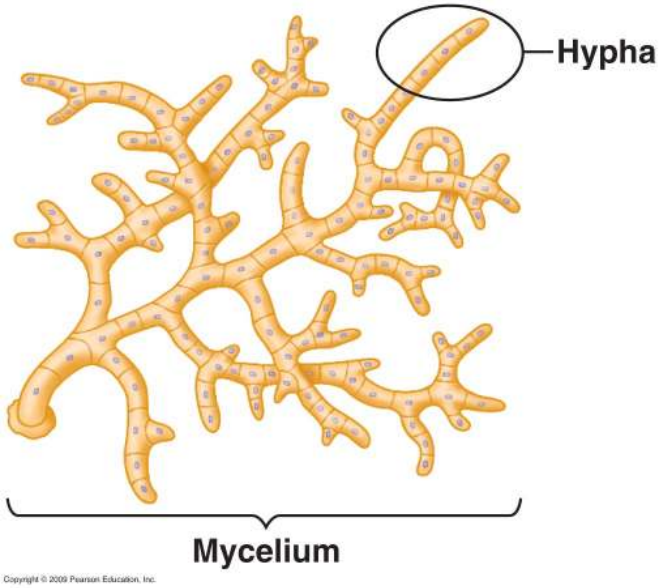
Budding

The nucleus of a fungal cell divides, and splits off from the rest of the cell by cytokinesis.

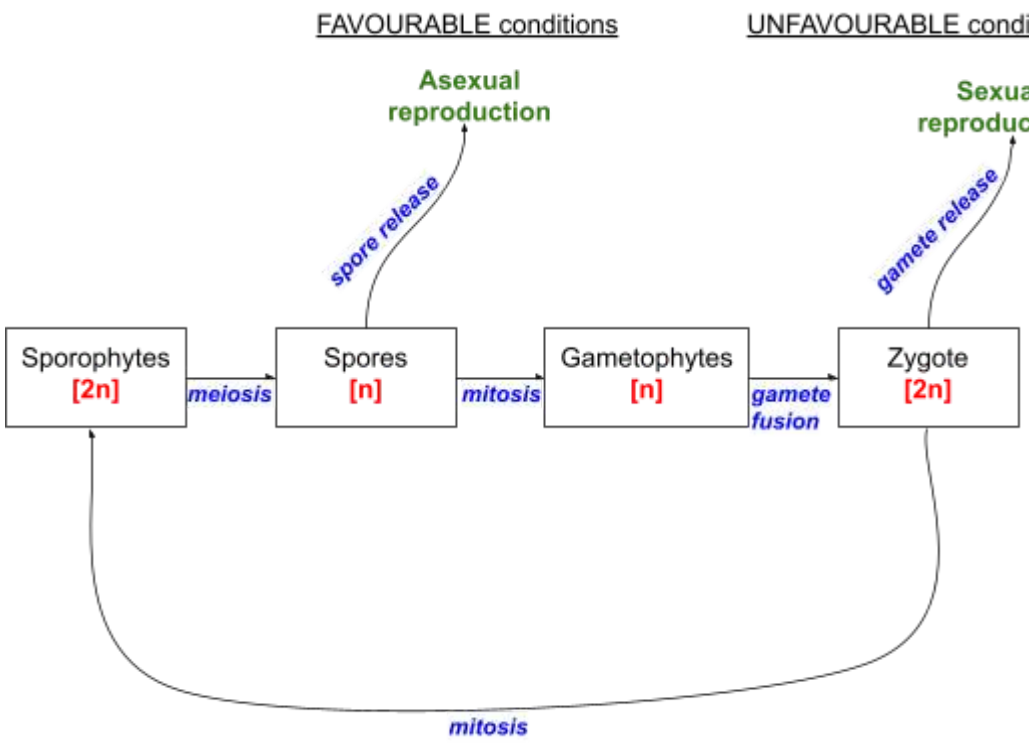


Complete budding: Separation → new cell

Incomplete budding: Bud stays attached to the mother → Keeps budding → hyphae → mycelium



Spores
Alternation of generation.



Fungi can reproduce both sexually and asexually within one lifetime.

1. Haploid spores are generated from diploid sporophytes through *meiosis*
2. If the conditions are <u>favorable</u>, these spores will be released into the environment
   → *Asexual reproduction*
3. If the conditions are <u>NOT favourable</u>, the spores will form into haploid gametophytes
   a. Both male and female gametes are produced
4. Fusion of these gametes (fertilisation) produces *zygotes*, which are released into the environment
   → *Sexual reproduction*

These zygotes can duplicate via mitosis, producing more sporophytes

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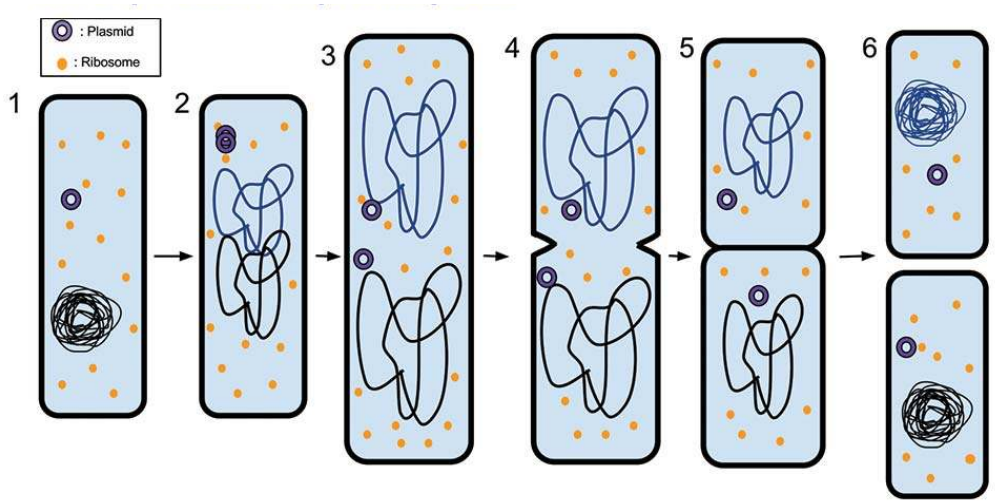
<u>IN BACTERIA</u>
Binary fission
A diploid (*2n*) mother cell splits into two diploid daughter cells which are genetically identical.

A cell grows, reaching the limit of its SA:V
↓
The organism replicates its nucleoid and plasmid DNA
(this ensures that each offspring retains a complete copy of the genome)
↓
The cytoplasm is pinched off at the area of cleavage
↓
A new cell wall is synthesised in this area



IN PROTISTS

Binary fission

In the same way as bacteria, but without a cell wall.

Budding

Only a small group of amoeba called *sarcodines* can bud.

A small outgrowth develops from the body of the parent sarcodine

↓

The offspring develop from this outgrowth and *completely separates* from the mother (no partial separation)

[5.1.2] Fertilisation, Implantation, Pregnancy and Birth in Mammals

CONTINUITY OF SPECIES

Reproductive mechanisms ensure the *success* of producing offspring and nurturing them to reproductive maturity. This is so they can in turn *reproduce and ensure the continuity of species through generations*.

Stage of development	Continuity of species
• Internal fertilisation	Ensures gamete fusion
• Implantation	Protects embryo
• Pregnancy	Provides embryo with nutrients
• Birth	Occurs after the foetus is fully developed to ensure survival
• Lactation	Sustains born offspring until it reaches reproductive maturity.

SEX HORMONES

Stages of sexual reproduction are regulated by hormones, ensuring reproductive success leading to the continuity of species.

They are responsible for:

- The development of reproductive organs
- The development of secondary sexual characteristics

In males	In females
➔ Deepened voice ➔ Thicker body hair, and more of it ➔ Increased size & thickness of muscles and bones	➔ Larger breasts ➔ Widening hips ➔ Growth of pubic hair

Types of sex hormones:

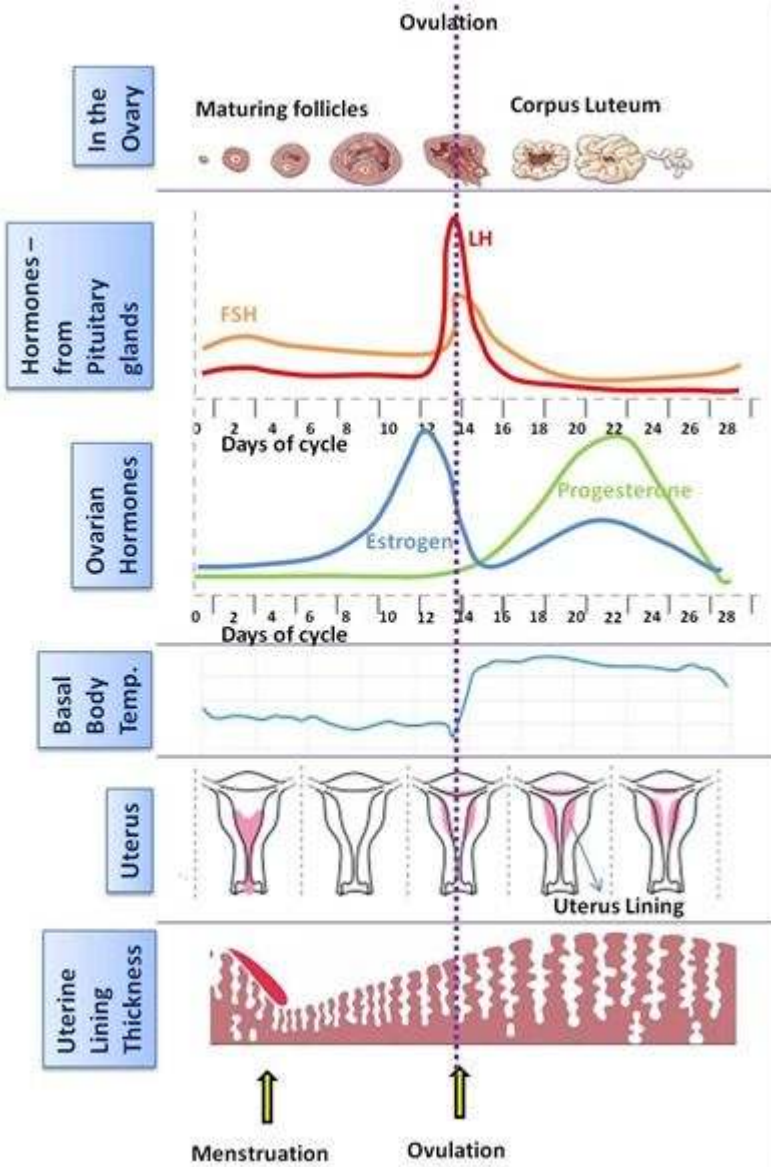
ANDROGENS	*Main male hormone, but also present in females.* The main androgen in males is called testosterone, produced by the testes. Androgen is responsible for the development of sperm, and secondary sexual characteristics in males.
OESTROGEN	*Main female hormone, but also present in males.* Involved with the development of the female reproductive system, and secondary sexual characteristics in females.
PROGESTERONE	*Only in females.* Ensures that the thickness of the endometrium is maintained during pregnancy.

MENSTRUATION AND OVULATION

Hormones involved

Hormone	Function	Location produced
1. Follicle stimulating hormone	Maturation of follicles	Pituitary gland (brain)
2. Luteinising hormone	Final maturation of follicles Development of corpus luteum	
3. Oestrogen	Development of sex organs	Ovaries
4. Progesterone	Controls pregnancy	

The process



FSH rises, causing follicle cells to develop

↓

FSH boosts oestrogen production (positive feedback loop)

↓

Graafian follicle released during ovulation

↓

Oestrogen stimulates LH secretion

↓

LH inhibits FSH and oestrogen production

↓

LH causes dominant follicle to rupture and release egg

↓

Follicle develops into corpus luteum

↓

Corpus luteum produces progesterone and oestrogen

↓

LH and FSH production inhibited by progesterone. Prevents 2nd egg being released

↓

Endometrium thickens

LH and FSH spike when follicle ruptures.
Progesterone spikes when the corpus luteum is largest.
Oestrogen spikes at both these times.

If fertilised	If NOT fertilised
Zygote implanted into endometrium ↓ Zygote produces Human Chorionic Gonadotropin (hCG) ↓ hCG maintains corpus luteum to produce progesterone for 12 wks ↓ The placenta develops fully and begins secreting progesterone ↓ Corpus luteum degrades, the placenta maintains the pregnancy	Corpus luteum degenerates ↓ Oestrogen and progesterone production stopped ↓ Endometrium is shed (menstrual flow) ↓ LH and FSH produced again ↓ Cycle repeats

GAMETOGENESIS

The formation of gametes from germinal epithelial cells.

Spermatogenesis

Germinal epithelium cells divide by mitosis (2n)
↓ mitosis
They grow into primary spermatocyte (2n)
↓ meiosis I
These split into 2 secondary spermatocytes (2n)
↓ meiosis II
Each spermatocyte produces 2 spermatids (n)
↓
[Spermatids are provided nutrition by sertoli cells]
↓ differentiation
Spermatids develop into spermatozoa (n)

A total of 4 spermatozoa are produced in this process.

Oogenesis

Germinal epithelial cells divide by mitosis (2n)
↓ mitosis
Grow into primary oocytes (2n)
↓ meiosis I (stop @ prophase I)
Primary follicle formed (2n)
↓
Menstrual cycle starts @ puberty
↓ meiosis I
Divides into secondary oocyte and a polar cell (n)
↓ meiosis II (mitosis)
Secondary oocyte divides into an egg and another polar cell (n)
↓
An egg is formed and develops. The 3 polar cells are discarded

FERTILISATION

Gamete fusion and the production of a zygote

The process

Sperm are attracted to the egg by chemical signals

↓

Sperm pushes through the follicle layer around the egg

↓

The first sperm to break through binds to the zona pellucida

↓

Acrosome reaction triggered

↓

Acrosome bursts and digests the zona pellucida, allowing the sperm to enter the egg

↓

Cell membranes of the sperm and egg fuse

↓

Cortical reaction triggered

↓

Enzymes from the cortical granule are released, which harden the zona pellucida to stop more sperm entering

↓

The two nucleuses of the egg and the sperm undergo mitosis

↓

The entire cell divides, moving ½ of the egg's chromosomes and ½ of the sperm's chromosomes into separate cells

↓

Zygote created

Breeding times

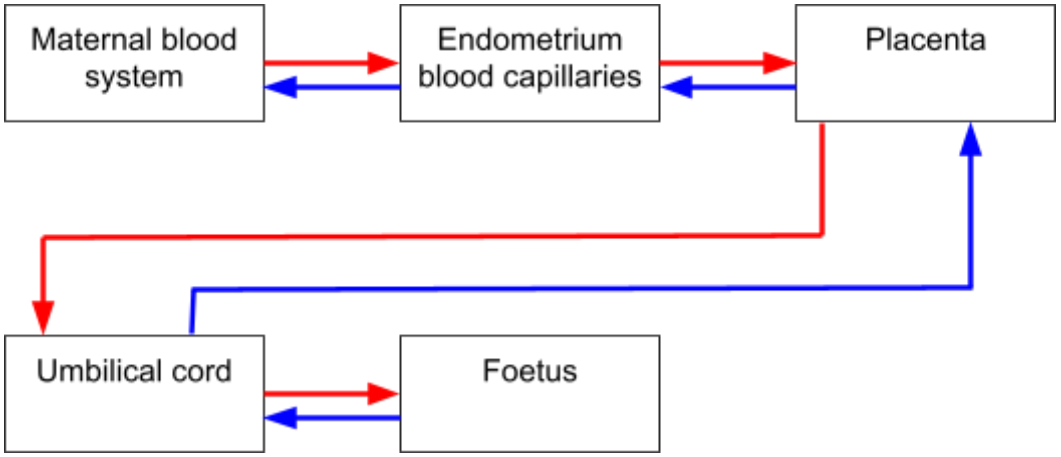
SEASONAL BREEDERS	CONTINUOUS BREEDERS
• The female animal is ready to mate at specific periods throughout the year Advantage: - The offspring are produced during favourable environmental conditions	• The female animal can mate at any time during the year due to the continuous reproductive cycle Advantage: - Offspring are continuously produced, ensuring population growth and continuity

IMPLANTATION

The embryo is embedded into the uterus.

The placenta

The foetus develops an umbilical cord and placenta.
The placenta is a disk-shaped tissue which gets embedded into the uterus wall by placental villi. It connects the blood systems of the mother and the foetus without them fusing.



Maternal → Foetus	Foetus → Maternal
- Oxygen - Glucose - Water - Minerals - Antibodies - Hormones	- Carbon dioxide - Urea - Water - Hormones

PREGNANCY

Human development

Zygote	Morula	Blastocyst	Embryo	Foetus
Fertilised egg	16 unspecialised cells	Mass of cells - Inner forms embro - Outer forms placenta	8wks after fertilisation	Embryo develops bone tissue. +8wks to birth

Hormonal control of pregnancy

Embryos produce human chorionic gonadotropin (hCG). hCG prevents the corpus luteum from degrading, thus stimulating progesterone and maintaining a thickened endometrium.

After 12 weeks, the placenta is fully developed. It takes over the production of progesterone and oestrogen, so the corpus luteum degrades.

BIRTH

When the offspring moves into the external environment from the mother's reproductive system.

Process of birth

The cervix relaxes and widens
↓
amniotic sac bursts, releasing fluid
↓
baby comes out
↓
placenta is expelled (afterbirth)

Hormonal control of birth

Progesterone prevents contractions in order to maintain a stable environment for the growing foetus.

In the last ⅓ of pregnancy, the placenta produces estriol.

Estriol inhibits the production of progesterone, while promoting oxytocin production.
Furthermore, the rise in oestrogen increases the number of oxytocin receptors in the endometrium.
Oxytocin causes the uterine muscles to contract.

Uterine contractions begin slowly, and increase due to positive feedback, becoming stronger until the baby is pushed out.

LACTATION

The production of milk to sustain the offspring after birth.

Hormonal control of lactation

The hormone prolactin stimulates the mammary glands to produce milk.

- ➔ High levels of oestrogen before birth stimulate the production of prolactin (*positive feedback*)

- ➔ High levels of prolactin inhibit milk production (*negative feedback*)

Milk production will only begin with the decline of prolactin. This happens with the decline of oestrogen (ie. After the birth).

[5.1.3] Manipulation of Plant and Animal Reproduction in Agriculture

Scientific understanding of the mechanisms of reproduction allowed for the development of reproductive technologies.

Reproductive technologies are any technologies which increase the rate of reproduction, ensure survival and thus continuity of species.

Biotechnology is the manipulation of living organisms for human benefit.

WHOLE-LEVEL MANIPULATION

Selective breeding (plants & animals)

Method:

Humans breed together parents with desirable traits so that at least one of these traits appears in the offspring.

Breeding can be:

- ➔ Intraspecific (between the same species)
- ➔ Interspecific (between different species)

Advantages:	**Disadvantages:**
● If there are desirable traits in the male *and* the female, the offspring will most likely also have at least one of these traits	● Cannot guarantee the desired trait will be expressed ● Reduces the genetic diversity of a population ● Offspring from interspecific breeding are sterile as their gametes are not viable, thus the trait can not be passed on further

CELL-LEVEL MANIPULATION

Artificial insemination (animals)

Method:

1. Semen is collected from a male
2. A sexually receptive female is detected
3. Insemination performed by using an insemination gun to shoot semen into the female's cervix

Advantages:	**Disadvantages:**
• Able to synchronise births • Ability to choose the mates increases likelihood of passing down favourable traits • Ensurance of successful pregnancy increases yields	• Cannot guarantee the desired trait will be expressed • Reduced genetic variation

Artificial pollination (plants)

Method:

1. Pollen (male gamete) removed from the stamen of one plant
2. This pollen is applied to the stigma of another plant
3. Pollen fertilises the ovum

Advantages:	**Disadvantages:**
• Cross-breeding of favourable traits • Decreases likelihood of self-pollination • Ensures successful pollination of all plants, increasing the yield	• Cannot guarantee the desired trait will be expressed • Reduced genetic variation

Cloning (plants & animals)

Method:

1. Somatic body cell of organism A is removed, and the nucleus is extracted
2. This nucleus is put into the denucleated egg of a donor
3. The egg is implanted into a surrogate mother
4. The offspring is a clone of A

Advantages:	**Disadvantages:**
• Cloned individuals have identical requirements • The desired trait is guaranteed to be expressed	• Clones have no adaptability • Expensive • Ethical concerns • Clones have questionable health and life expectancy

GENE-LEVEL MANIPULATION

Transgenesis (plants & animals)

Method:

1. A gene for a specific trait is removed from a species
2. The transgene is held in recombinant plasmid
3. The gene is injected into the zygote of another species
4. The zygote + transgene is implanted into a surrogate mother
5. The offspring express the transgene in their phenotype

Advantages:	**Disadvantages:**
● Guaranteed expression of desired trait ● Increase yield ● Reduces use of harmful chemicals ● Increases genetic diversity ○ (as long as the transgenic organisms are not cloned)	● Transgenic organisms have low adaptability ● Mixing of GM organisms with wild populations ● Trade issues with non-GMO countries ● Unknown side effects on humans ● Transgenic organisms are usually cloned, decreases genetic variation

IMPACT OF REPRODUCTIVE TECHNOLOGIES

Initially, due to manipulation of reproduction, there are increased variations in a species.

However, since the offspring with favourable traits are then cloned, variations decrease again.

	gene level		species level		ecosystem level
↓ variations	→	↓ genetic diversity	→	↓ species diversity	→	↓ long-term biodiversity

This limits the adaptability of a species to changing selection pressures, therefore limiting the continuity of species.

5.2: Cell Replication

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

Genetic material refers to DNA.

Because DNA replication produces genetically identical offspring, all members of a species have the same genome (collection of genes) but different variations of that gene (alleles).

This means a species is defined by its genome.

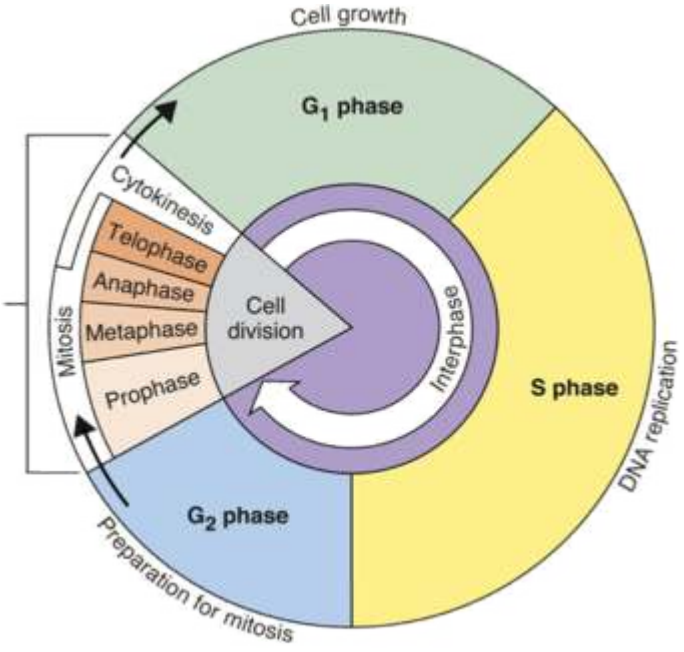
➔ No two species have the same genome

Therefore, exact DNA replication is important as it ensures that the correct genes are passed on from parent to offspring, so that the species can continue in the next generation.

DNA replication →	Inheritance of identical genes →	Inheritance of exact genome →	Continuity of species

[5.2.1] The Processes Involved in Cell Replication

THE CELL CYCLE

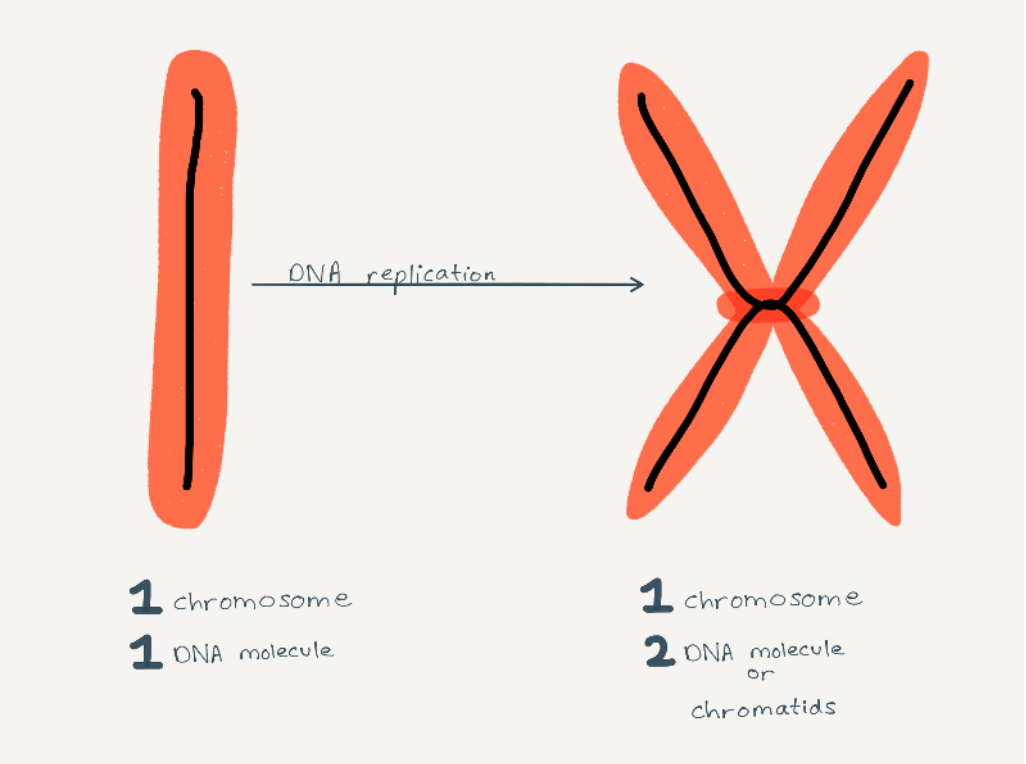


G_1 : growth phase	Cell grows ↓ Volume increases faster than SA ↓ SA:V decreases ↓
S : synthesis phase	DNA synthesis & replication ↓ Mutations ↓
G_2 : Growth phase 2	Detection of mutations ↓ Correction of errors ↓
Nuclear division	Prophase, metaphase, anaphase, telophase ↓
Cytoplasmic division	Cytokinesis

BEHAVIOUR OF CHROMOSOMES

Before replication: Many genes → 1 DNA → 1 chromosome
After replication: Many genes → 2 DNA → 1 chromosome

DNA replicates, chromosomes don't



When 2 DNA are attached at the centromere, they are known as 2 sister chromatids.

MITOSIS

Location: Somatic (body) cells.

Result: Diploid (*2n*) parent cell creates two diploid daughter cells.

Function: Growth, repair, maintenance.

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 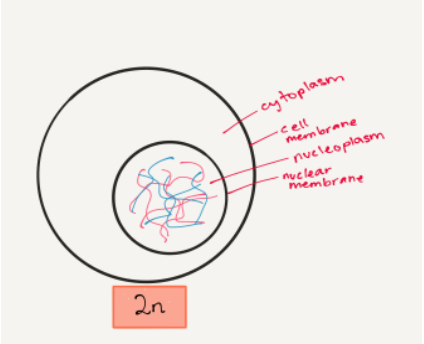
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Chromosomes can not be seen individually, they are called chromatin material.

They are thin, long and coiled together.

DNA replication takes place

46 chromosomes DNA replication → 46 chromosomes
46 DNA 92 DNA/ chromatids
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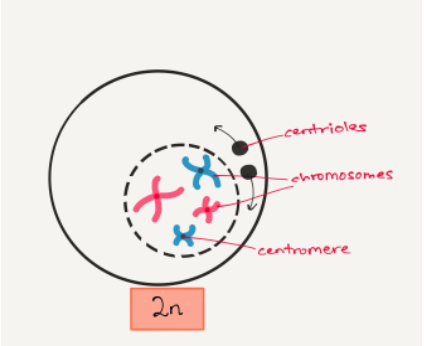
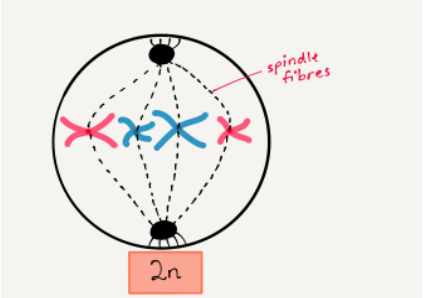
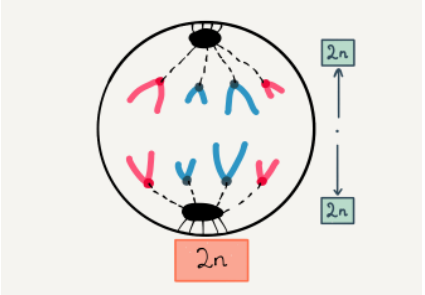
 
</td>
<td>

Nuclear membrane dissolves.
Centriole divides and the halves move towards opposite poles of the cell.
Individual chromosomes become visible.

Each is made of 2 chromatids due to DNA replication in interphase.

</td>
</tr>
<tr>
<td>METAPHASE
 
</td>
<td>

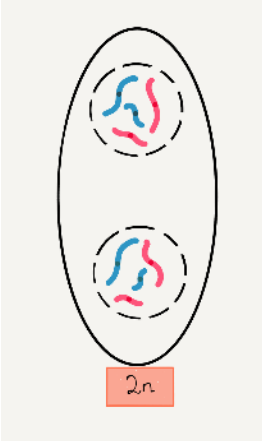
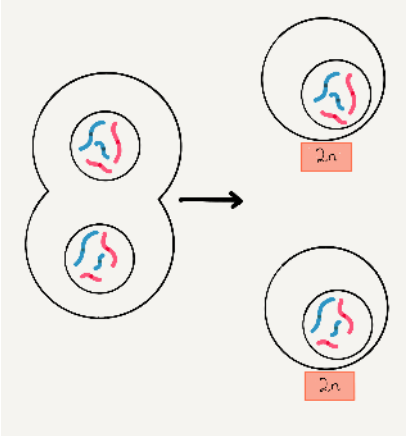
Chromosomes align across the equator of the cell in a single file.
Spindle fibers form which connect the centrioles to the centromeres of the chromosomes.

</td>
</tr>
<tr>
<td>ANAPHASE
 
</td>
<td>

Spindle fibers contract.
Chromosomes with one DNA are pulled to opposite poles of the cell.

They are no longer chromatids since the DNA molecules are not connected.

</td>
</tr>
</table>

TELOPHASE  	• Nuclear division completes. • Nuclear membrane forms around the chromosomes at each pole. • 2 diploid, identical nuclei are formed.
CYTOKINESIS  	• Cytoplasm cleavages • Cytoplasmic division • 2 diploid, identical daughter cells are derived.

MEIOSIS

Location: Germline (sex) cells.

Result: Diploid parent cell creates 4 haploid (*n*) daughter cells.

Meiosis I: *Reduction division*. Creates haploid cells from diploid cells.

Meiosis II: Maintains haploid number in daughter cells.

Function: Gamete production for sexual reproduction.

↓

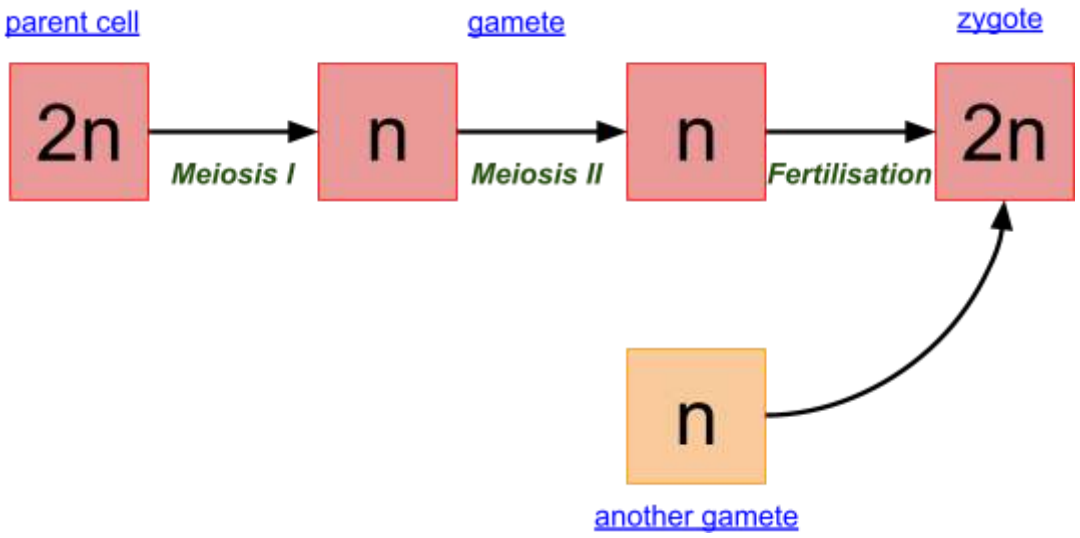
Ensures inheritance of the genome

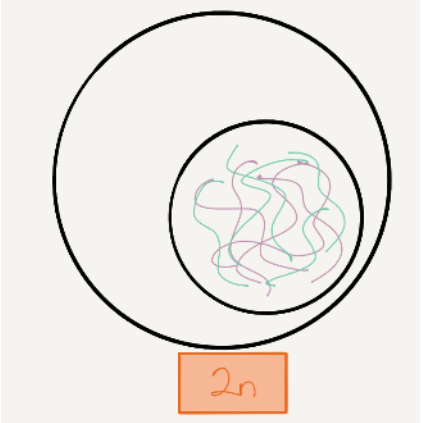
↓

Source of variation

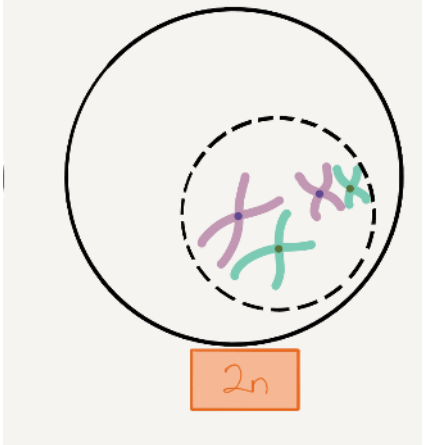
↓

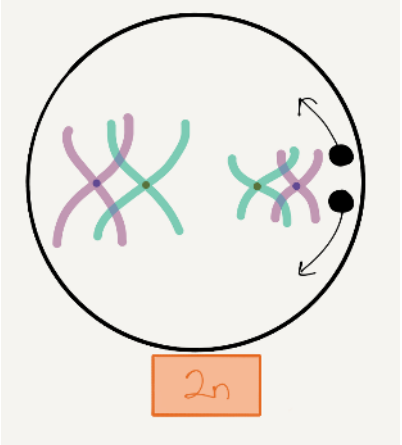
Continuity of species



INTERPHASE  	• Chromosomes can not be seen individually, they are called chromatin material. ○ They are thin, long and coiled together. • DNA replication takes place *46 chromosomes* → *46 chromosomes* (DNA replication) *46 DNA* → *92 DNA/ chromatids*

PROPHASE I

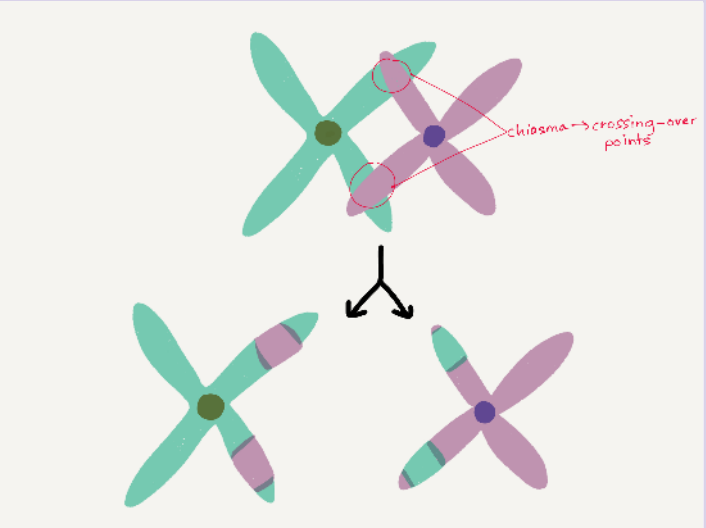




- Chromosomes with 2 chromatids appear.
- Chromosomes are arranged in homologous pairs.
- Nuclear membrane dissolves.
- Centrioles move towards the poles.

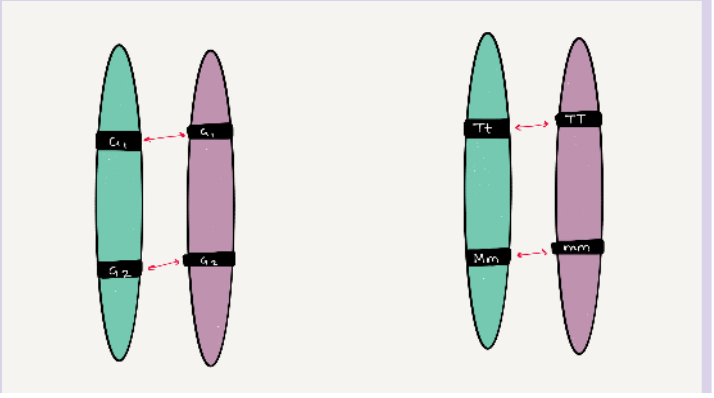
Crossing over

- Non-sister chromatids cross over and exchange maternal and paternal genes across the chiasma.
  - Creates **unique combinations of allele pairs on the same chromosome**
  - This recombination of maternal and paternal genes causes variation



Alleles are variations of the same gene. Alleles for a common trait are found on the same locus of a chromosome.

- If a segment does not cross over in the exact area, mutations occur.



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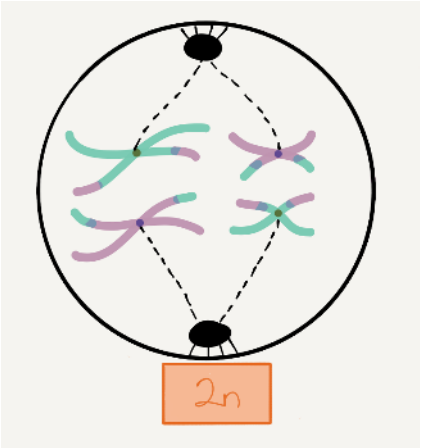
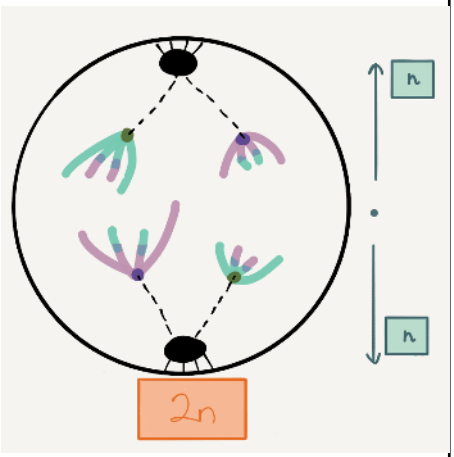
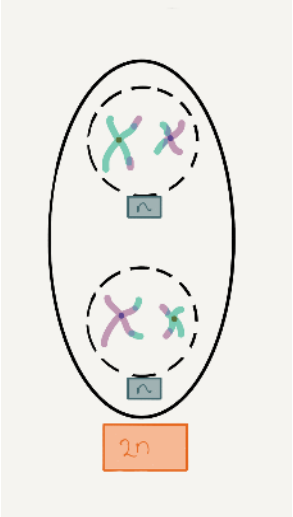
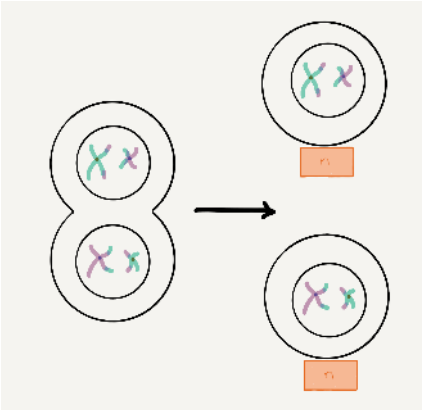
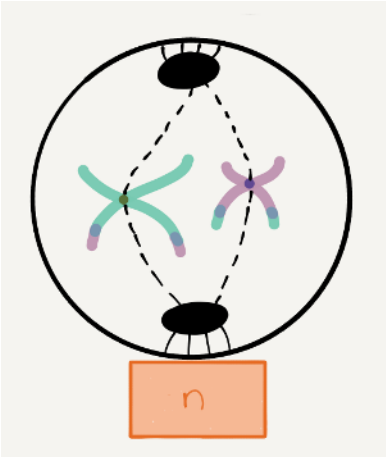
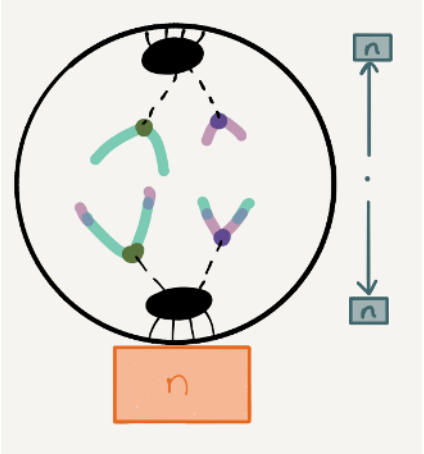
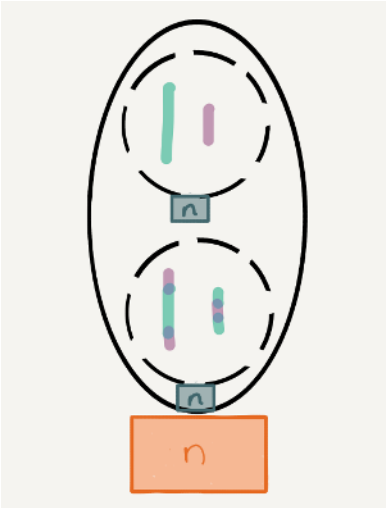
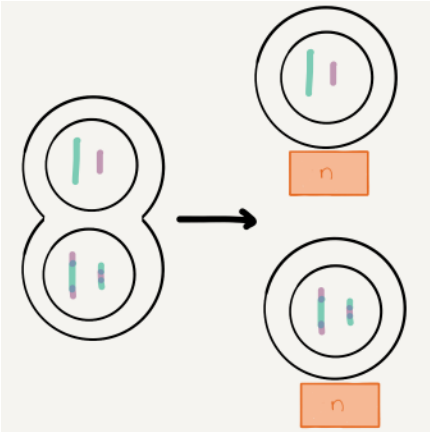
<td>METAPHASE I
 
</td>
<td><u>Independent assortment</u>
• Homologous pairs of chromosomes line up in double file along the cell's equator.
• The arrangement is random, and independent of other chromosomes.

<u>Random segregation</u>
• Chromosomes of the homologous pairs move to the poles independently of each other.

Create a unique combination of alleles in the gametes.

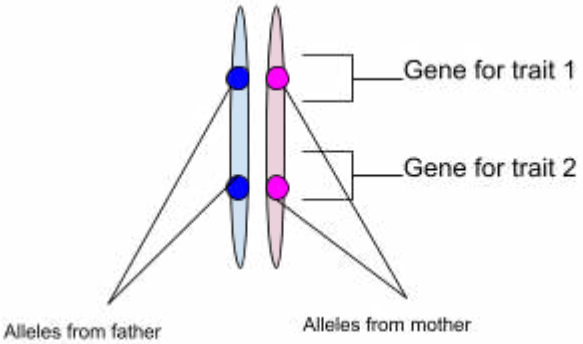
Crossing over, independent assortment and random segregation are a source of genetic variation.</td>
</tr>
<tr>
<td>ANAPHASE I
 
</td>
<td>• Spindle fibers contract and pull chromosomes to the opposite poles.</td>
</tr>
<tr>
<td>TELOPHASE I
 
</td>
<td>• Nuclear division completes.
• 2 haploid, non-identical nuclei are formed.</td>
</tr>
</table>

CYTOKINESIS I 	• Cytoplasmic division completes. • 2 haploid, non-identical cells are formed. • Meiosis I completes.
PROPHASE II	N/A
METAPHASE II  	• Chromosomes line in a single file along the equator of the cell.
ANAPHASE I  	• Spindle fibers contract, pulling chromosomes to opposite poles.

TELOPHASE II  	• Nuclear division completes. • 2 haploid non-identical nuclei are formed per daughter cell.
CYTOKINESIS II  	• Cytoplasmic division completes. • 2 haploid, non-identical gametes are formed per daughter cell. • In total, 4 haploid, non-identical gametes are formed.

When fused, the offspring will have 2 chromosomes in each pair, one containing **alleles from the father** and the other containing **alleles from the mother**.

Offspring's genotype



Furthermore, gametes are randomly fertilised, allowing for **different, random alleles to create a genotype** for a trait in the offspring. This increases genetic variation within the offspring.

Mitosis vs. Meiosis

	MITOSIS	MEIOSIS
Type of cell produced	Somatic	Gamete
No. of cells produced	2	4
Location	Somatic cells	Germline cells
Function	Growth and repair	Gametes for sexual reproduction
No. of chromosomes per daughter cell	Diploid $2n \rightarrow 2n$	Haploid $2n \rightarrow n \rightarrow n$
Degree of variation between parent cell and daughter cells	Identical (Excluding variations due to mutations)	Large variation (due to random segregation and crossing over of genes)
DNA replication	Yes, in interphase.	Yes, in interphase.

WATSON AND CRICK DNA MODEL

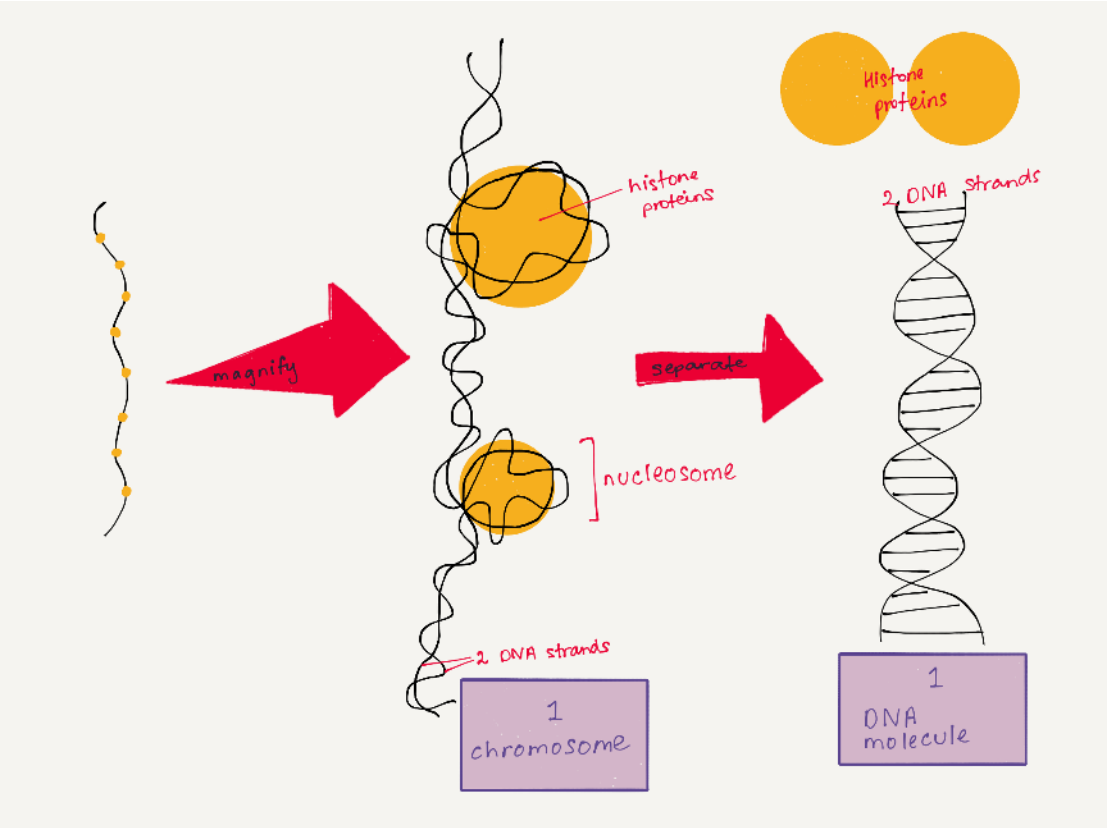
History of the DNA model

The W+C model of DNA was built in 1953.

- ➔ It was a double helix structure
- ➔ Scientists involved: James Watson, Francis Crick, Rosalind Franklin, Morris Wilkins

Watson and Crick	vs.	Franklin and Wilkins
• Watson was a biologist • Crick was a physicist • Perfect communication • Analytical skills		• Franklin was an x-ray crystallographer • Wilkins produced DNA crystals • Franklin found angles of diffraction using crystalised DNA to create a model • Limited communication

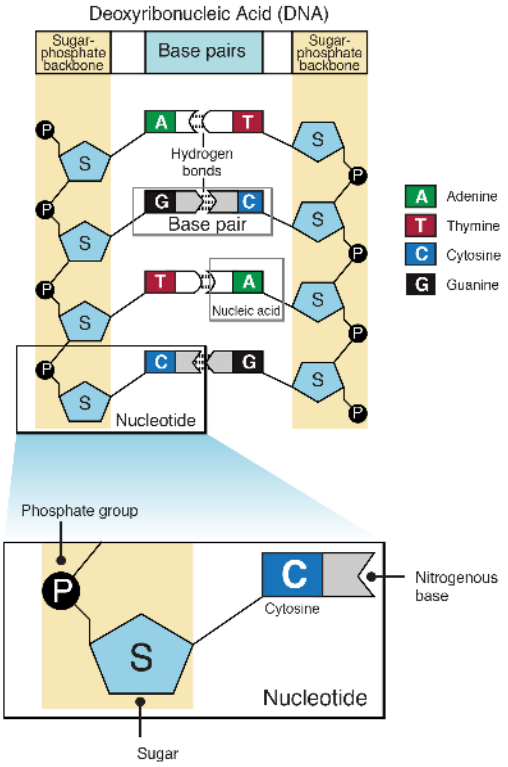
Nucleotide composition, pairing and bonding



DNA and chromosomes have a 1:1 ratio since a chromosome is a molecule of DNA wrapped around histone proteins.

Many genes → 1 DNA → 1 chromosome

DNA is a 3D molecule made of a sequence of nucleotides. Unwrapped, it looks like this:



Nitrogenous base + phosphate + sugar
=
Nucleotide

The nitrogenous bases are:

1. Adenine (A)
2. Guanine (G) → Purines

3. Thymine (T)
4. Cytosine (C) → Pyrimidines

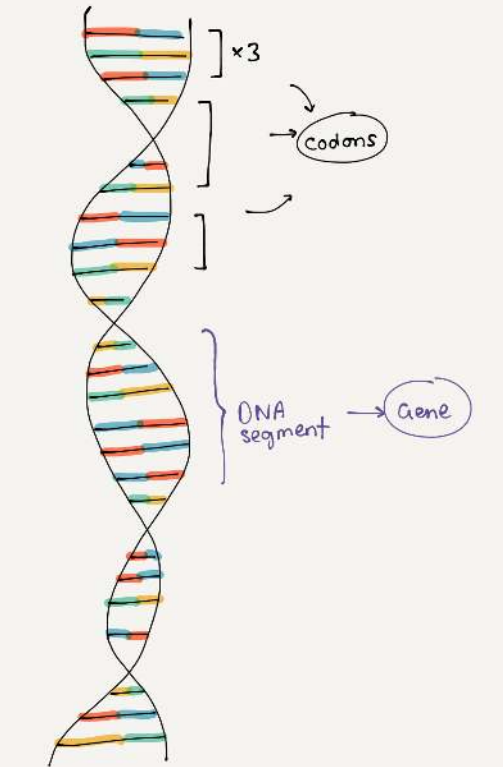
They undergo the complementary base pairings:

A = T [2 hydrogen bonds]

G ≡ C [3 hydrogen bonds]

Gene expression

The process by which the genotype shows the phenotype is called gene expression. It happens through protein synthesis.



GENE (DNA segment)
↓
Sequence of nucleotides/ nitrogenous bases
↓
Sequence of codons
↓(each codes for an amino acid)
Sequence of amino acids
↓
Polypeptide chain
↓(one or more chains link)
Specific protein
↓
Specific trait/ characteristic

This is how each gene determines a trait.
Many genes ⇒ many proteins ⇒ many traits

DNA REPLICATION IN EUKARYOTES

DNA replication occurs in the synthesis stage of interphase.

DNA REPLICATION

Importance

DNA replication → identical DNA produced → cell replication

↗ Mitosis → DNA shared (same generation)

↘ Meiosis → DNA inherited (next generation)

↓

Passing genome on

↓

Recombination of genes

↓

Genetic variations

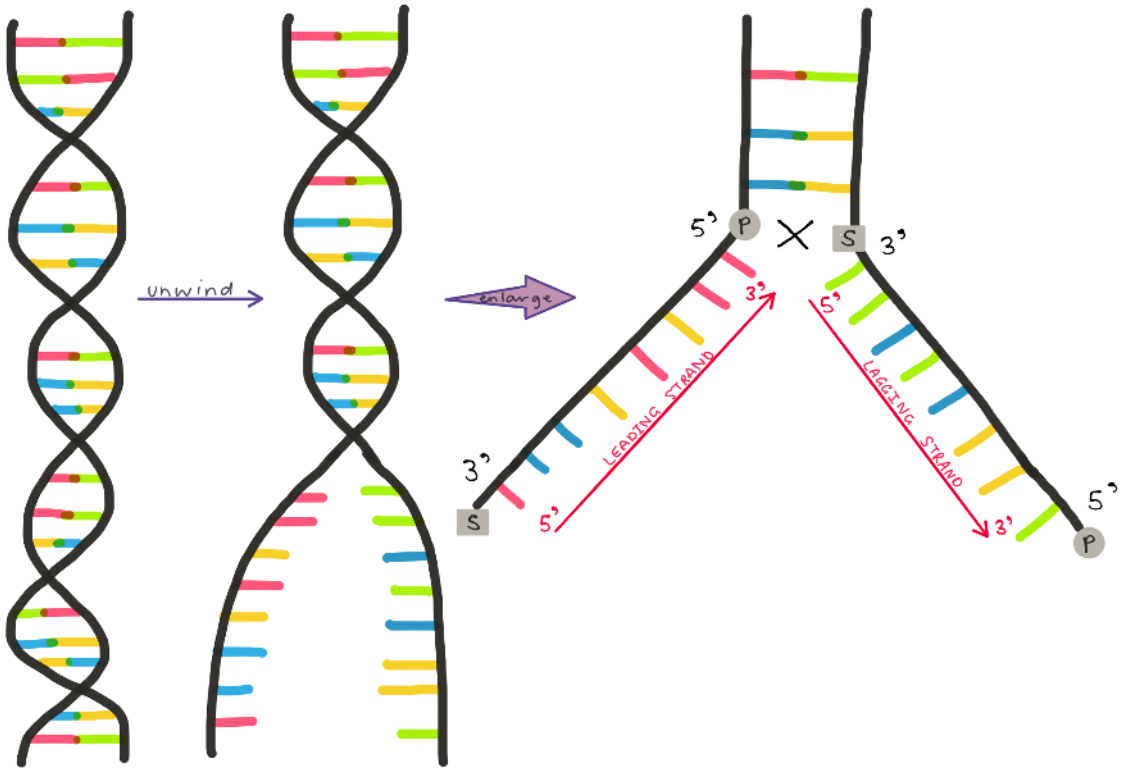
↓

Continuity of species

The original strand

1 molecule of DNA has 2 complementary strands which go from 3'→5' or 5'→3' .

- ➔ 3' ⇒ ends in sugar
- ➔ 5' ⇒ ends in phosphate



The strands are antiparallel.

- The *leading strand* forms continuously from 5'→3' towards the replication fork
- The *lagging strand* forms in fragments from 5'→3' away from the replication fork

The process of DNA replication

INITIATION: Ligase relieves the strain on a twisted DNA helix

↓

Helicase uncoils and separates complementary strands

↓

ELONGATION

↙ ↘

<u>Leading strand</u>

DNA polymerase (III) adds nucleotides segments (primers) in the 5' → 3' *towards* the replication fork.

DNA polymerase also proofreads the strand to correct any base pair errors.

↓

Ligase seals the gaps between DNA

<u>Lagging strand</u>

RNA polymerase adds a RNA primer to the strand, allowing DNA polymerase to bind.

↓

DNA polymerase (III) adds Nucleotides next to the RNA primer in okazaki fragments.

↓

DNA polymerase (I) replaces The RNA primer with DNA and corrects any pairing errors.

↓

Ligase seals the gaps between DNA

↓

The RNA primer moves to the next area, and the cycle repeats until the entire lagging strand is formed.

↓

Strands recoil into a double-helix shape.
2 identical daughter strands produced.

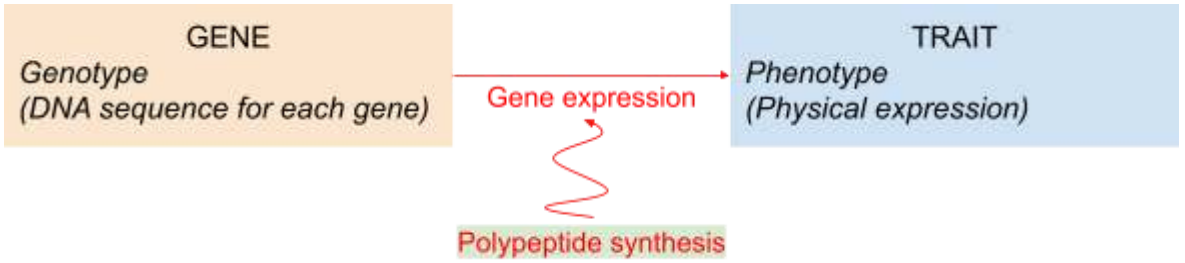
↓

TERMINATION

The 2 new DNA strands each contain 1 old template strand and 1 new strand. This is called <u>semi-conservative replication</u>.

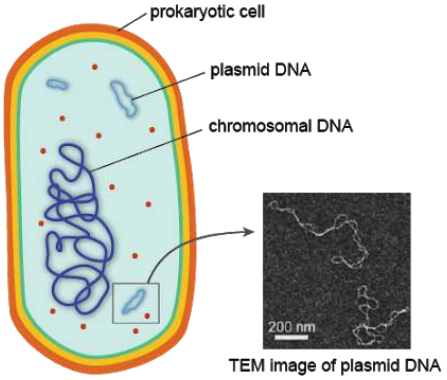
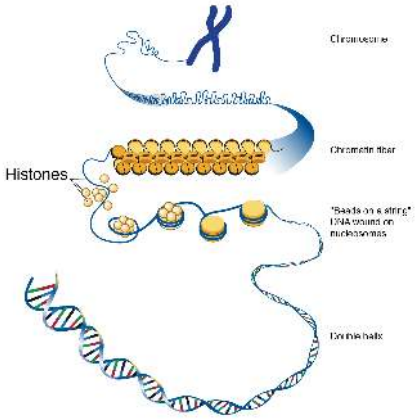
5.3: DNA and Polypeptide Synthesis

Why is polypeptide synthesis important?



Polypeptide synthesis is important because it allows for gene expression to occur so that organisms can live.

[5.3.1] DNA in Prokaryotes vs. Eukaryotes

	Prokaryotes	Eukaryotes
DNA shape	Chromosomal DNA in tight supercoils around scaffold protein. - For essential features Loop-like plasmid DNA. - For non-essential features  TEM image of plasmid DNA Conservative replication	Double helix coiled around histone proteins. - DNA wrapped around histones to make nucleosomes  Semi-conservative replication.
DNA location	Cytoplasm. - Naked	Nucleus - Membrane bound

NON-NUCLEAR DNA

Some organelles (eg. Mitochondria, Chloroplasts) have their own DNA.

Mitochondrial DNA (mtDNA) is identical through female lineage, and can be used to trace a genetic line.

- Evolves more rapidly, as replication is not checked by repair enzymes
- Abundant in every cell

DNA vs. RNA

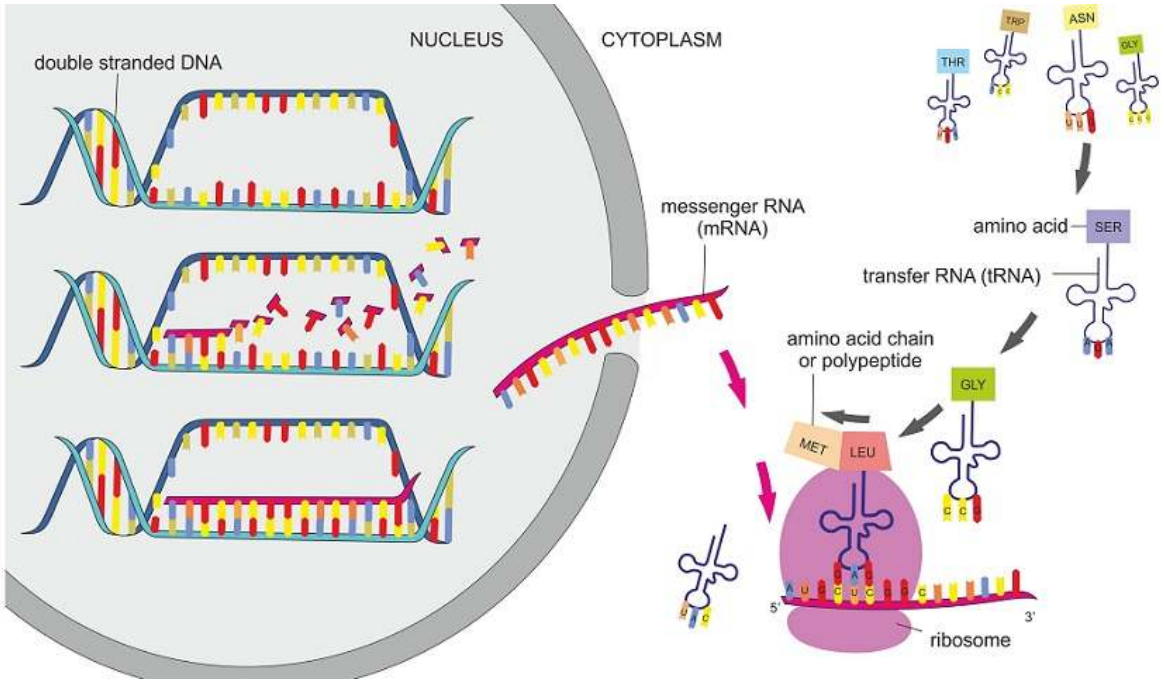
DNA *Deoxyribonucleic acid*	RNA *Ribonucleic acid*

- Double helix
- Deoxyribose sugar
- 4 bases
  - Adenine
  - <u>Thymine</u>
  - Guanine
  - Cytosine
- Stores genetic material
- Undergoes replication
	- Single helix
	- Ribose sugar
	- 4 bases
	  - Adenine
	  - <u>Uracil</u>
	  - Guanine
	  - Cytosine
	- Converts the information stored in DNA into proteins
	- Doesn't replicate

	mRNA ● Takes the coding sequence for specific proteins from DNA and transports to the ribosomes tRNA ● Carries amino acids to the ribosome. Specific types of amino acids correspond to the anticodon rRNA ● Form the ribosome. They move along the mRNA, and bind the tRNA

[5.3.2] <u>POLYPEPTIDE SYNTHESIS</u>

<u>Function</u>: To produce a polypeptide and then a protein
<u>Significance</u>: Allows gene expression to occur



Transcription

RNA polymerase enzyme unzips a segment of the DNA helix (a gene)

↓

There are 2 strands: Non-coding and coding.
To replicate the coding strand, mRNA forms against the non-coding strand.

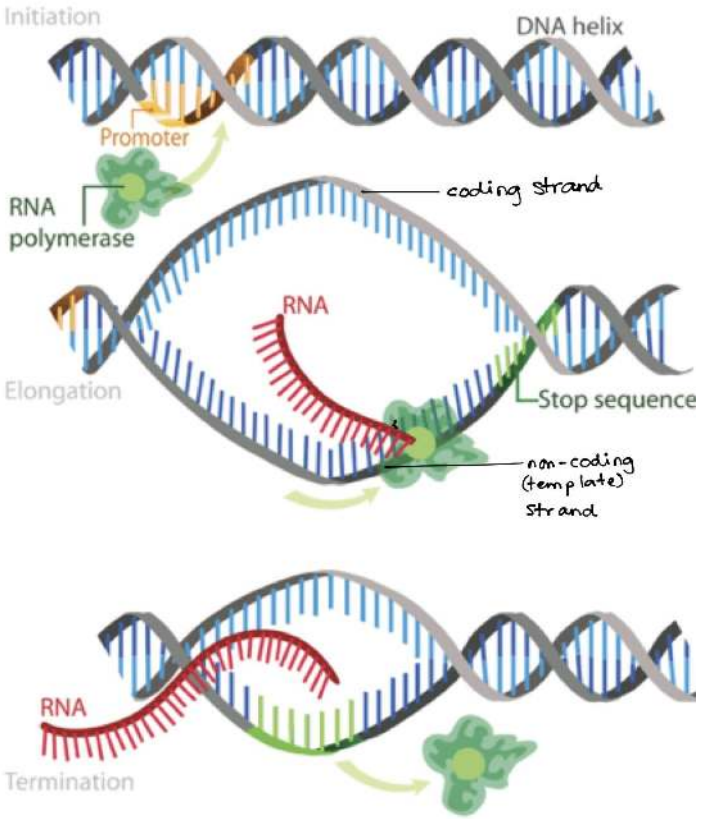
↓

Pre-mRNA is created when free-floating nucleotides attach in complementary pairs.

- Instead of thymine, RNA uses uracil

↓

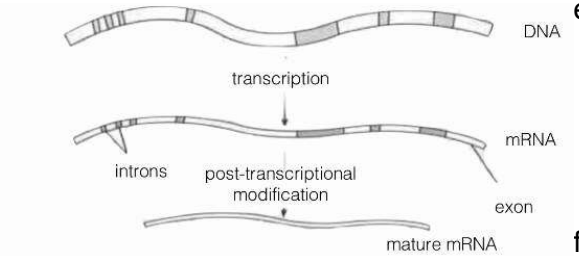
DNA rezips and never leaves the nucleus.



Post-transcriptional modification

The transcribed mRNA contains introns and exons.

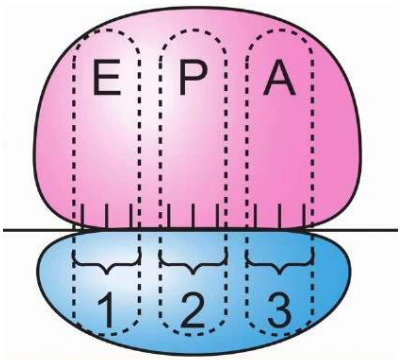
Exons ⇒ codes for amino acids
Introns ⇒ non-coding regions



Endonuclease enzymes cuts out the introns.
The exons are connected by ligase enzymes to form mature mRNA.

Translation

A ribosome is composed of a large and small subunit, with 3 binding sites: E, P, A.



the

Each codon (3 base pairs) of the mRNA has a complementary tRNA which carries an amino acid.

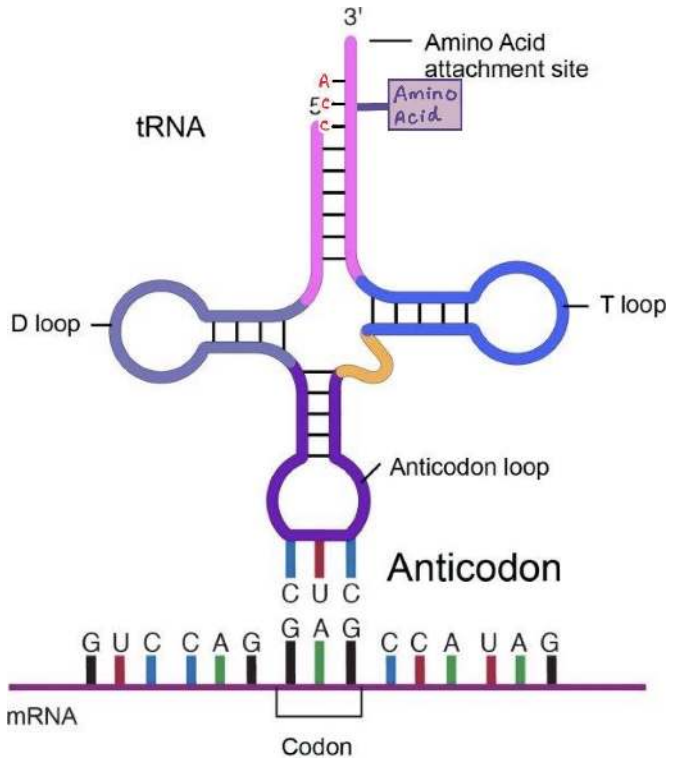
Each codon codes for a specific amino acid.

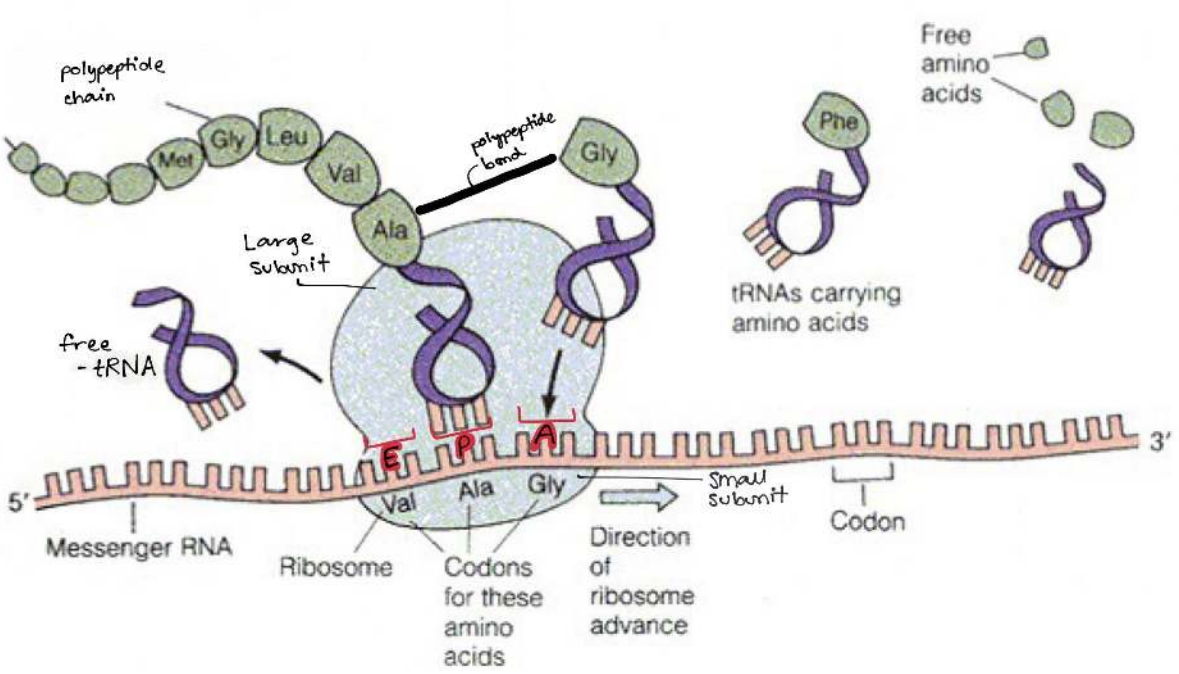
The order and number of amino acids determines the function of protein.

Second letter

First letter	U	C	A	G	Third letter
U	UUU Phe UUC Phe UUA Leu UUG Leu	UCU Ser UCC Ser UCA Ser UCG Ser	UAU Tyr UAC Tyr **UAA Stop** **UAG Stop**	UGU Cys UGC Cys **UGA Stop** UGG Trp	U C A G
C	CUU Leu CUC Leu CUA Leu CUG Leu	CCU Pro CCC Pro CCA Pro CCG Pro	CAU His CAC His CAA Gln CAG Gln	CGU Arg CGC Arg CGA Arg CGG Arg	U C A G
A	AUU Ile AUC Ile AUA Ile AUG Met	ACU Thr ACC Thr ACA Thr ACG Thr	AAU Asn AAC Asn AAA Lys AAG Lys	AGU Ser AGC Ser AGA Arg AGG Arg	U C A G
G	GUU Val GUC Val GUA Val GUG Val	GCU Ala GCC Ala GCA Ala GCG Ala	GAU Asp GAC Asp GAA Glu GAG Glu	GGU Gly GGC Gly GGA Gly GGG Gly	U C A G

1.





1. Initiation

   mRNA attaches to the small subunit. The starting codon has the sequence AUG

   ↓

   A complementary initiator tRNA [1] (carrying methionine) attaches to the A site and moves to the P site

   ↓

   The tRNA [2] of the next codon lands on the A site

   ↓

   A polypeptide bond is created between the two amino acids

   ↓

   tRNA [1] drops it amino acid and moves to the E site as a free tRNA

2. Elongation

   The initiation tRNA is recycled in the cytoplasm

   ↓

   The tRNA [2] now carries 2 amino acids and moves to the P site

   ↓

   The ribosome continues to read the mRNA

   ↓

   tRNA [3] lands on the A site, and a polypeptide bond is created between the amino acids of tRNA [2] and [3]

   ↓

   tRNA [2] drops its amino acid and moves to the E site, giving tRNA [3] 3 amino acids

   ↓

   This cycle continues as the polypeptide chain elongates

3. Termination

   The ribosome reaches a stop codon

   - UAG
   - UGA
   - UAA

   The stop codon doesn't code for a protein, but it releases the polypeptide chain from the P site by releasing chemicals.

MUTATIONS

If a mutation occurs during DNA replication, when transcribed and translated it will lead to an incorrect amino acid in the polypeptide chain, creating an abnormal protein.

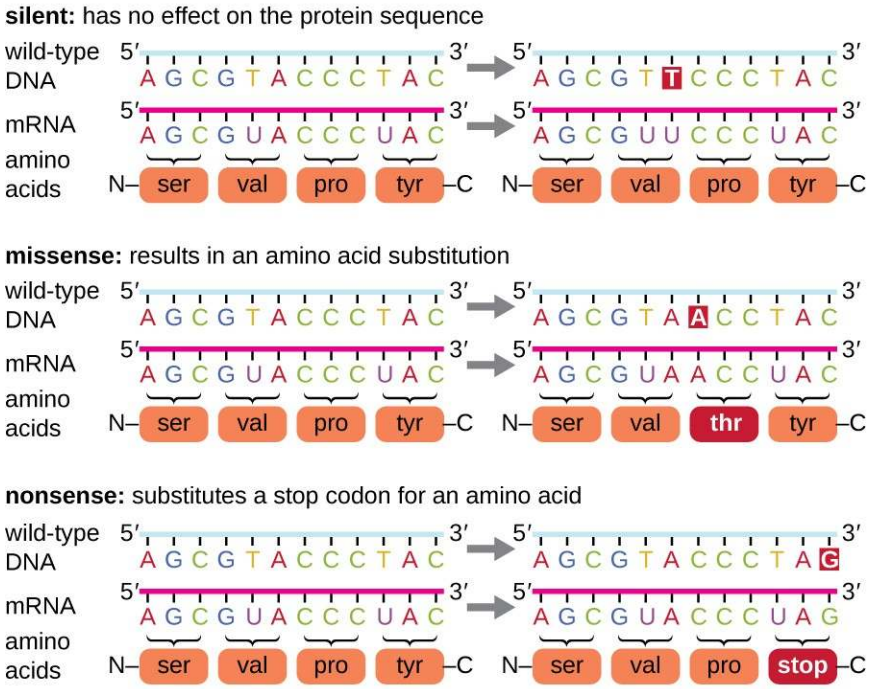
This will impact the metabolism and thus the health of the organism.

Base substitutions

1 base of the coding strand is replaced with an incorrect, non-complimentary one.

This may:

- ➔ Cause no change → silent mutation
- ➔ Change 1 amino acid → missense mutation
- ➔ Shorten the chain → nonsense mutation



Base insertions and deletions

Bases are removed or added.

↓

This changes the reading frame, as bases are read in 3 (codons)

↓

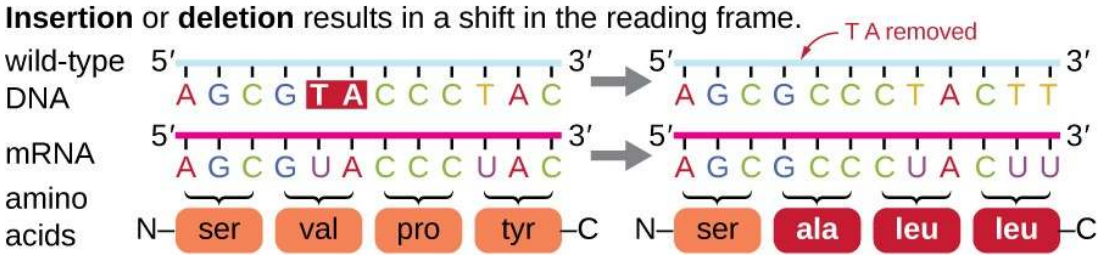
Frame-shift mutation

↓

Significantly changes the amino acid sequence

↓

Non-functional protein

➔ 

GENES AND THE ENVIRONMENT ON PHENOTYPE

Although the genotype is expressed as the phenotype, traits may be masked or changed by the environment.

Genes + Environment = Phenotype

The alteration may be temporary (usually) or permanent.

Genes will only express traits if the environment is suitable.

Examples:

- *Having tall genes will not make an individual tall if there is not enough nutrients in the environment.*

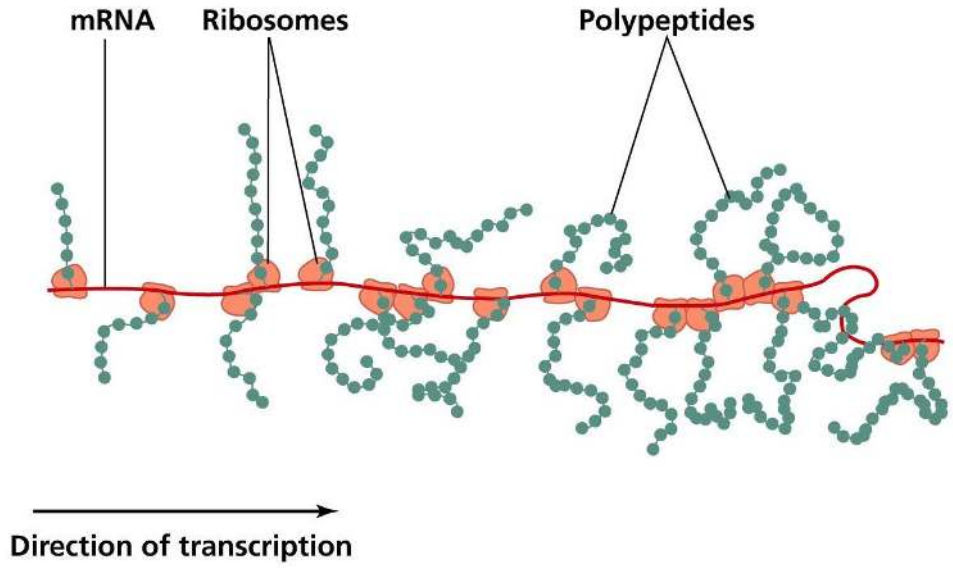
- *The petals of hydrangea plants change colour based on soil pH*
  acidic ⇒ blue
  alkaline ⇒ pink

- *Siamese cats will grow dark colours on the tips of their bodies when the temperature decreases, and lose them when it increases.*

[5.3.3] Investigate the Structure and Function of Proteins in Living Things

Polyribosomes

Multiple ribosomes can form parts of the polypeptide chain, which come together to form the complete chain.

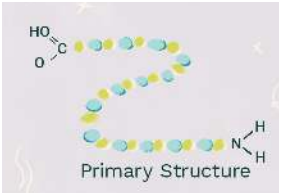
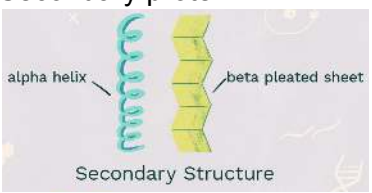


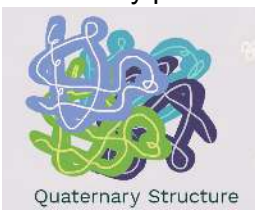
Protein conformation

One or more polypeptide chains can be structurally modified in the golgi apparatus to form specific proteins.

Amino acids (monomers) → polypeptides → proteins (polymers)

Types of proteins

PROTEIN SHAPE	FEATURES
Primary protein 	- A single, linear chain of amino acids
Secondary protein 	- A linear sheet of amino acids - Pleated into alpha helices or beta sheets

Tertiary protein 	- A linear sheet of amino acids - Pleated into alpha helices or beta pleated - Folded into a 3D structure
Quaternary protein 	- 2 or more proteins (of any structure) come together to form a single protein

Once shaped, proteins are held together by hydrogen bonds.

Each structure leads to the other, meaning that each protein will show characteristics of the shape before it.

Types of proteins and functions

TYPE OF PROTEIN	FUNCTION OF PROTEIN
Neurotransmitters [primary /secondary]	***Transport.*** Chemically transfer electrical impulses across the synapsis.
Lysozymes [secondary]	***Storage.*** The fluid within the lysosome organelle.
Enzymes [tertiary]	***Biocatalysts.*** Speed up biochemical reactions.
Haemoglobin [quaternary]	***Transport.*** Transports oxygen
Hormones [quaternary]	***Messengers.*** Chemical messengers which coordinate cell activity
Antibodies [quaternary]	***Immunity.*** Flights pathogens for immunity
Collagen [quaternary]	***Growth & repair.*** Muscle fiber which strengthens muscles

5.4: Genetic Variation

How can the genetic similarities and differences within and between a species be compared?

Genetic similarities and differences can be compared at a genetic level within species and at a species level between species.

Within a population (intraspecific) :
- ➔ Similarities show common ancestry
- ➔ Differences show genetic diversity due to genetic variation

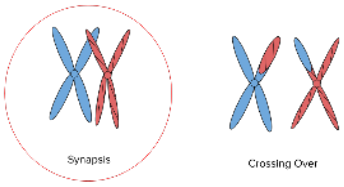
Between populations (interspecific) :
- ➔ Similarities show evolutionary relationships
- ➔ Differences show species diversity due to species variation

[5.4.1] Sources of Variation

Genome: The complete set of genes in a species
Gene pool: The complete set of alleles (gene variations) in a species.

Genetic diversity: total of all the genetic characteristics in the genetic makeup of a species.

Sources of genetic variation:

Source	Effect
Crossing over *(prophase)* 	Homologous chromosomes exchange genes, creating **new combinations of alleles** in the chromosome.
Independent assortment *(metaphase I)* & Random segregation *(anaphase I)*	Homologous chromosomes arrange themselves and separate independently, leading to **random allele combinations** in the 4 non-identical gametes.
Random fertilisation	Non-identical male and female gametes fuse to create an offspring with a **unique combination of alleles** to form unique genotypes.
Mutations	Produce **new alleles** to increase the possible variations.

Meiosis + Fertilisation + Mutations = recombination of genes

↓

Genetic variation

↓

Favourable adaptations

↓

Natural selection

↓

Reproductive maturity of fittest organisms

↓

Inheritance of favourable variations

↓

Genetic isolation

↓

Speciation (new species)

↓

Diversity

- Genetic diversity → *variations in gene combinations*
- Species diversity → *variations of species in an area*
- Biodiversity → *variations of living things in an area*

↓

Evolution

Therefore, genetic variation leads to evolution. (Neo-Darwinism).

[5.4.2] Mechanisms of Inheritance

Chromosomes

- **Autosome**: contains genes responsible for body traits (skin, hair, eye colour)
  1-22nd homologous pairs in humans.

- **Sex-chromosomes**: contains genes for gender and secondary sexual characteristics
  23rd pair in humans
  - *XX (females)*
  - *XY (males)*

Every body cell contains a complete set of **homologous chromosome pairs**.
In humans, there are 23 pairs in total.

MUTATIONS
Mutations occur in both the somatic and germline cells.

Mutations in somatic cells → expression in individual → no inheritance

Mutations in germline cells → not expressed in individual → gametes → inheritance → expressed in offspring

Gametes carry genetic variation, meaning that a trait will only be passed down if it occurs in the gametes. **Inheritance only occurs in germline cells**.

AUTOSOMAL INHERITANCE

Inheritance on germline cells where the genes inherited are on the 1st - 22nd pairs of chromosomes, and code for body characteristics.

In a specimen, there is a **gene** for each trait, and 2 variations of this gene known as **alleles**.

- ➔ The alleles can be dominant or recessive.
- ➔ Dominant alleles are always shown over recessive ones.

Eg. G_T is the gene for height.
T and t are the alleles (tall and short)
T is dominant, t is recessive.

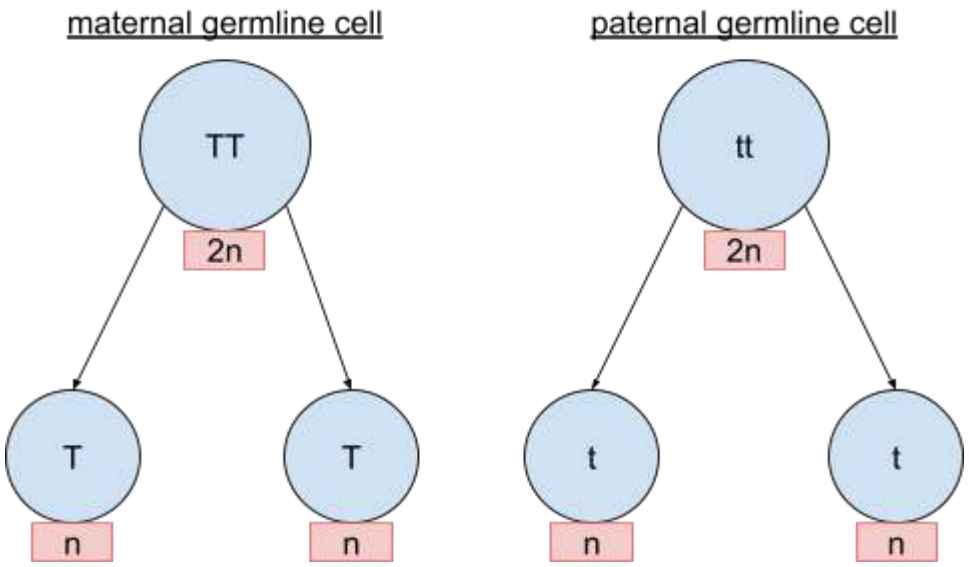
P1: Parental generation

The alleles of both parents

Pure tall x Pure short
TT x tt

G1: Gametes

Since only ½ of each parent's genetic information is passed onto the offspring, they only have allele from each parent.



F1: Filial generation 1

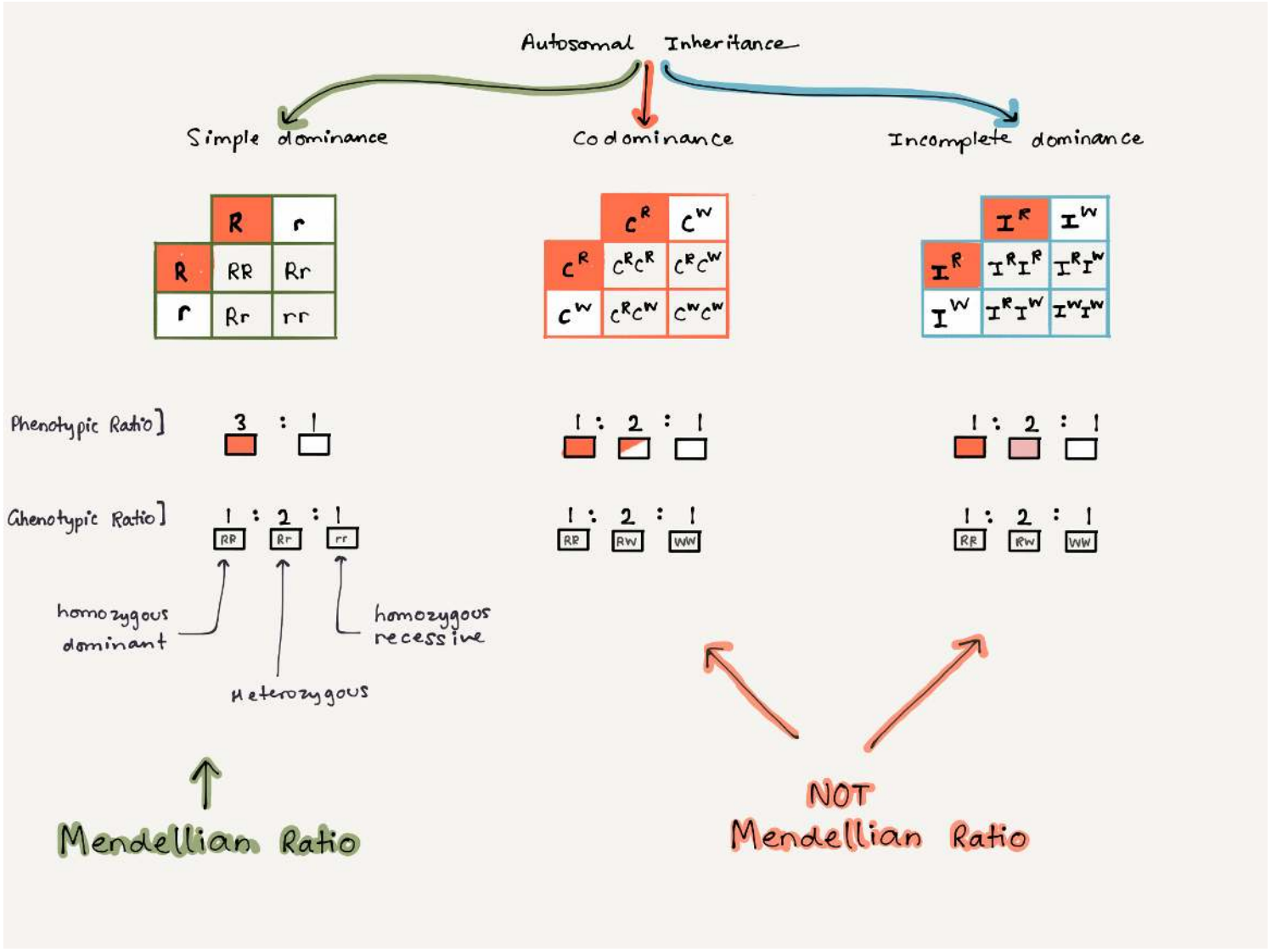
	T	*T*
t	*Tt*	*Tt*
t	*Tt*	*Tt*

Offspring genotype: 100% heterozygous.
Offspring phenotype: 100% tall.

Simple dominance: One allele is completely dominant over the other.

Codominance: Both alleles are dominant, so they both show <u>without blending</u>.

Incomplete dominance: Both alleles are dominant, so they are <u>blended</u> in the phenotype.



Multiple alleles:
While there are only 2 alleles per genotype in an individual, some traits can be coded for more than 2 alleles.
(Aka. **Polygenic traits**)

If there are many variants, it is known as continuous distribution.
If there are few variants, it is a discrete distribution.

Example: Blood groups
There are 3 alleles that code for blood types: A, B and O

- O is recessive
- A and B are both dominant over O, but equally dominant over each other

Therefore, there are 4 blood groups for the genotypes created:

$I^A \rightarrow$ A
$I^B \rightarrow$ B
i $\rightarrow$ O

Genotype	Alleles	Phenotype
I^AI^A	I^A	Blood type A
I^Ai	I^A and i	
I^BI^B	I^B	Blood type B
I^Bi	I^B and i	
ii	i	Blood type O
I^AI^B	I^A and I^B	Blood type AB

SEX LINKAGE

Inheritance on germline cells where the genes inherited are on the 23rd pair of chromosomes. These genes code for gender and secondary sexual characteristics.

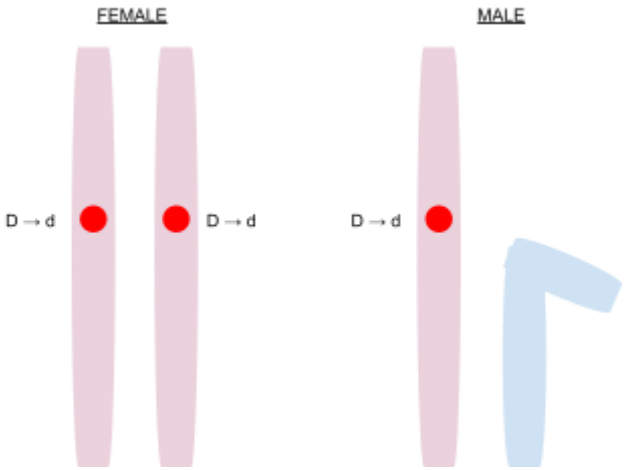
Due to a mutation, an autosomal gene can be transferred to the X chromosome of the 23rd (sex) pair.

↓

It becomes an **X-linked gene**.

↓

It undergoes **sex-linked inheritance**



$X^D > X^d > Y$

Female gametes → XX
Male gametes → XY

- Never carried on the Y chromosomes
- Only carried on the X chromosomes

X-chromosomes can be dominant or recessive, whereas Y chromosomes are always recessive.

➔ Therefore, a female can be a carrier
➔ Males can never be a carrier.

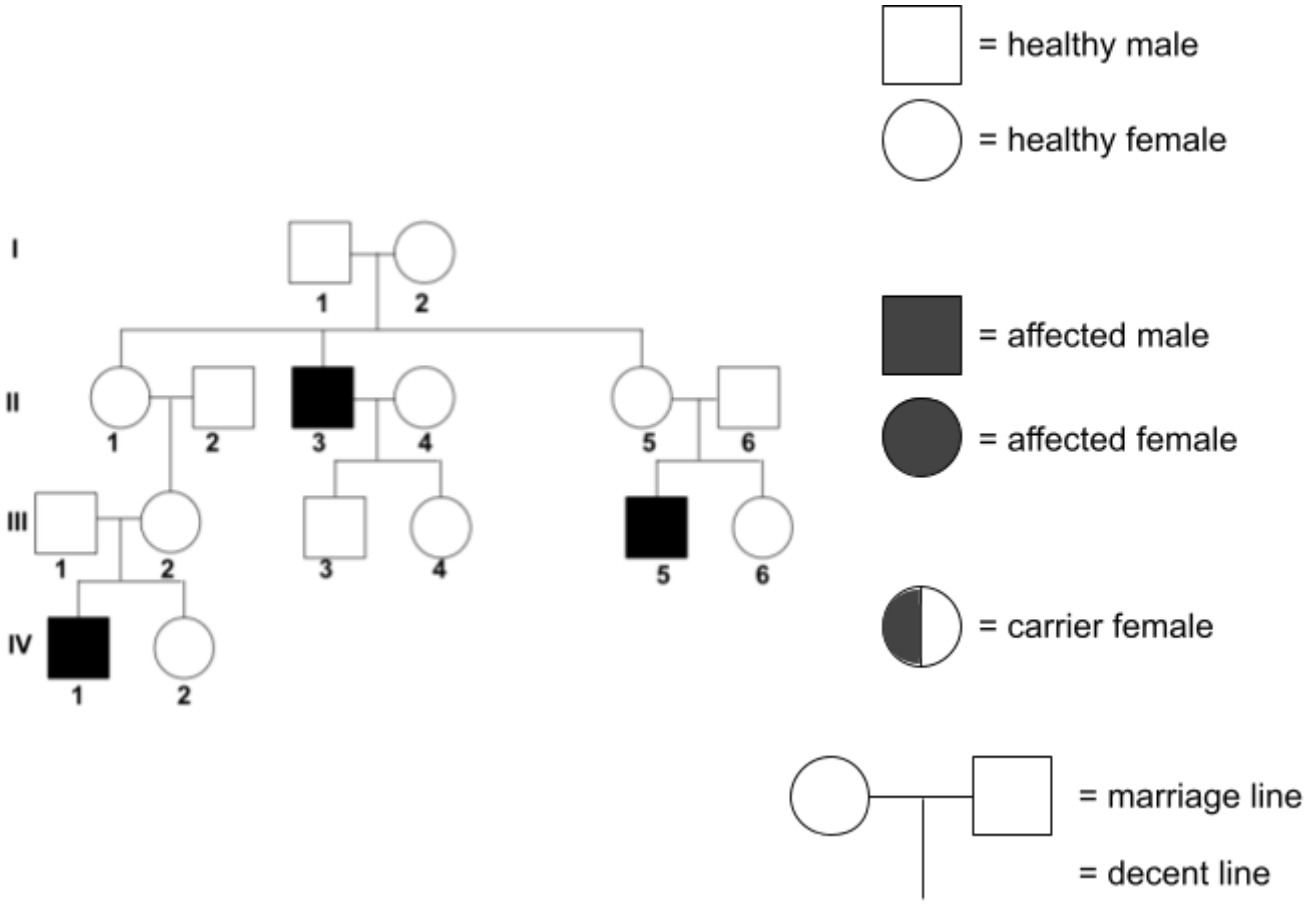
If the allele for a sex-linked disease is recessive (e):

X^E = dominant f. allele (most dominant)	X^e = recessive f. allele	Y = m. allele (most recessive)

Genotype	Alleles		Gender	Phenotype
X^EX^E	X^E		Female	Normal
X^eX^e	X^e		Female	Diseased
X^EX^e	X^E	X^e	Female	Carrier
X^EY	X^E	Y	Male	Normal
X^eY	X^e	Y	Male	Diseased

PEDIGREES

Graphical representations of inheritance patterns of a trait in related individuals over generations.



Patterns of inheritance

Autosomal dominant	➔ Males and females affected	
Autosomal recessive	➔ Males and females affected	
X-linked dominant	Male with trait: ➔ Passes onto all daughters ➔ Passes onto no sons	Female with trait: ➔ Passes to both daughters and sons
X-linked recessive	Male with trait: ➔ All daughters are carriers	Female with trait ➔ All sons are affected

[5.4.3] Population Genetics

The study of frequency of traits/ characteristics in a population.

↳ Genotype
↳ Phenotype
↳ Alleles

Changes in frequency → Change to population → Genetic variation → Evolution

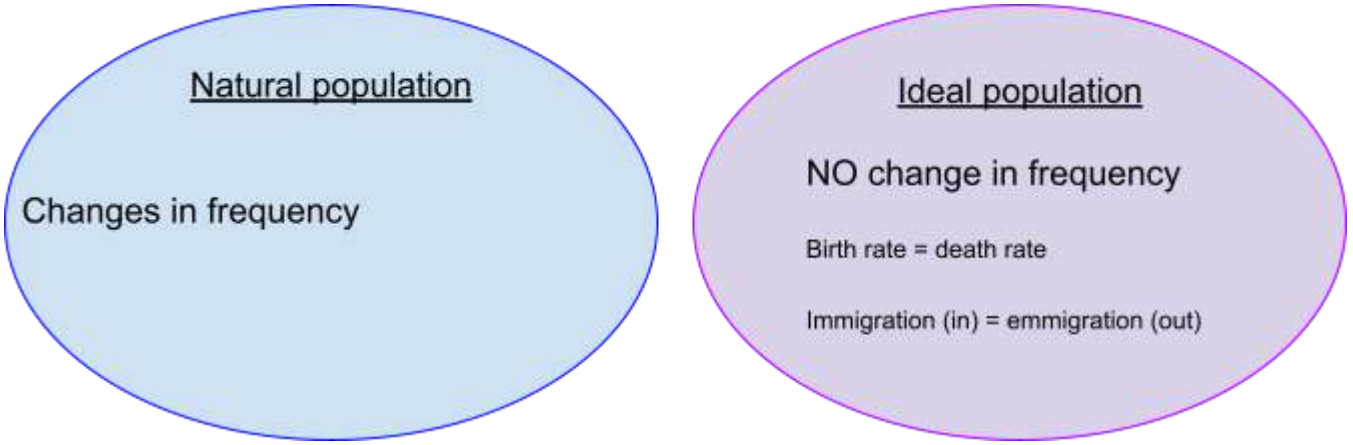
Studying population genetics allows us to:

- Predict the survival of a species through genetic variation
- Conservation management
- Predict the inheritance of diseases and disorders in a population
- Trace evolution

Mathematical model for frequency

$$Frequency\ of\ specific\ allele/\ genotype/\ phenotype\ = \frac{No\ of\ that\ allele/\ genotype/\ phenotype.}{Total\ no.\ of\ alleles/\ genotypes/\ phenotypes}$$

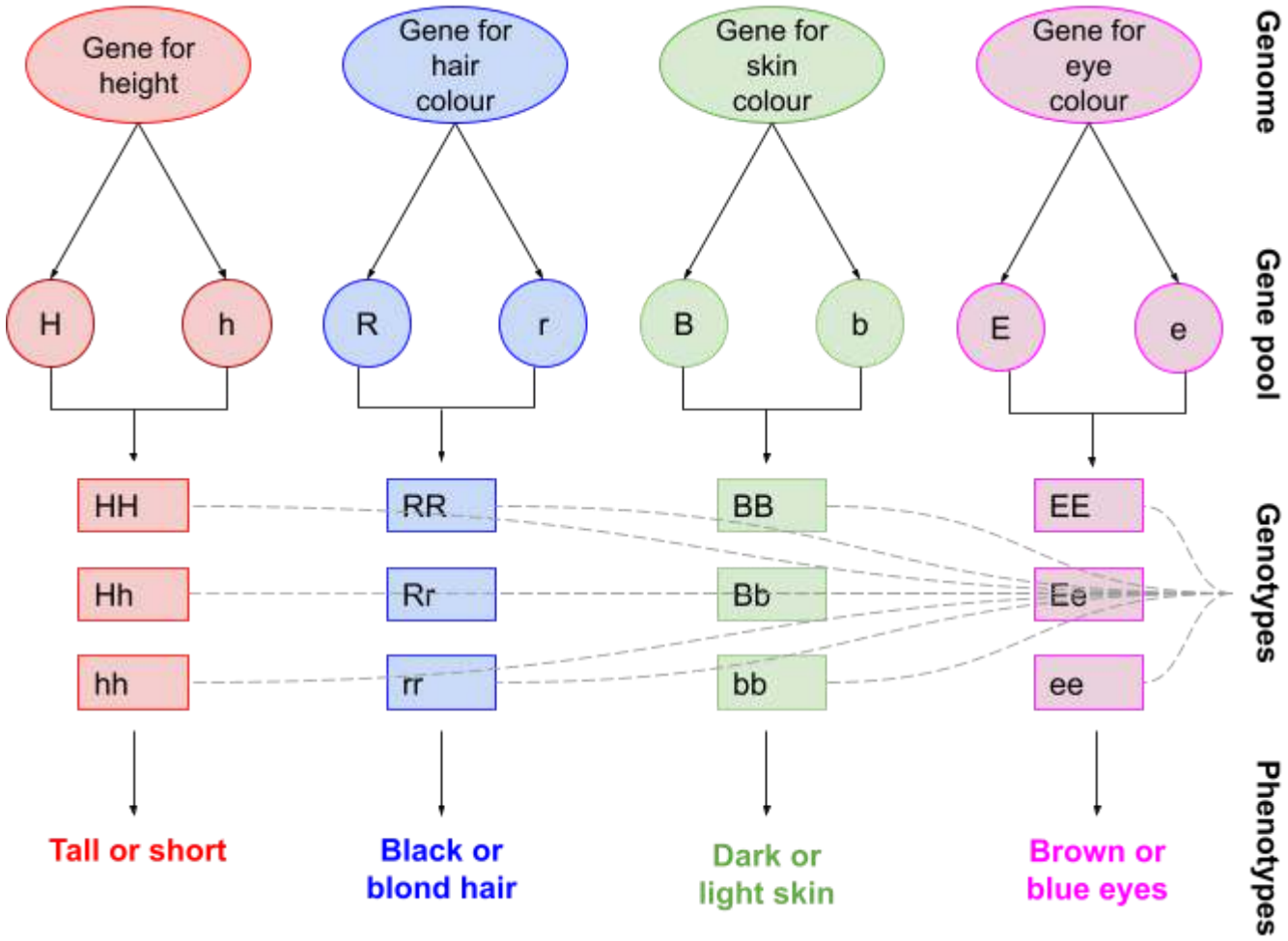
Modelling populations



Changes in a natural population can be identified when compared to an ideal population.

➔ Trends, patterns and limitations in the frequency data can be identified

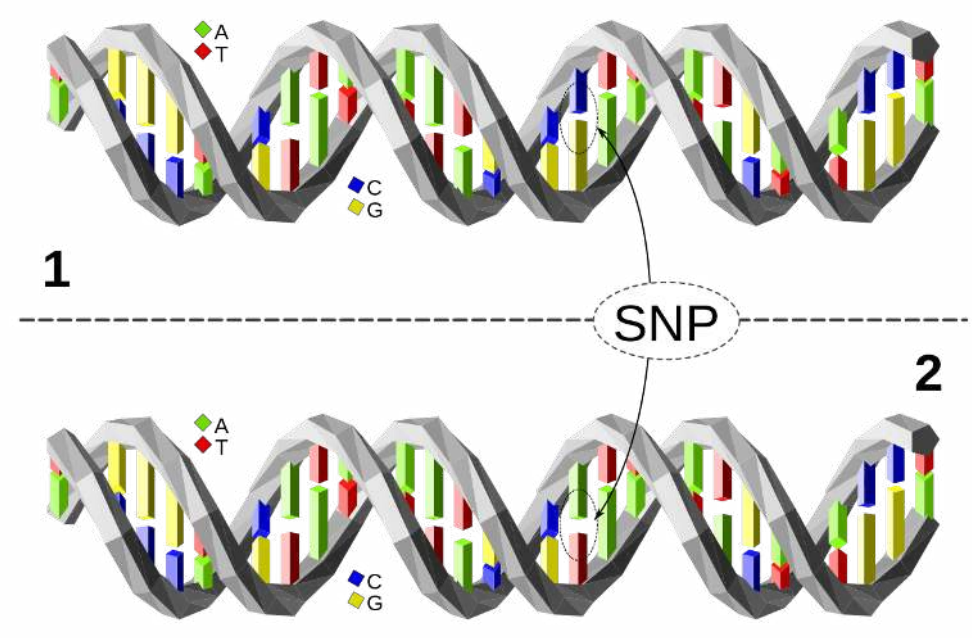
Population genetics: Shows how the gene pool changes over time.
Genetic diversity: The total of all genetic characteristics in a species.
Genetic variability: The tendency for genetic traits in a population to vary between individuals.



Genome	A collection of *all* genes responsible for *all* traits. The <u>same</u> for all members of a population.
Gene pool	Each gene has different variations or alleles. Collection of all the alleles in a population.
Genotype	Every individual inherits 2 alleles per trait. Each pair of alleles is a genotype. The genotype is <u>different</u> between members of a population.
Phenotype	Each genotype codes for 1 trait. The phenotypic expression of each genotype (or pair of alleles).

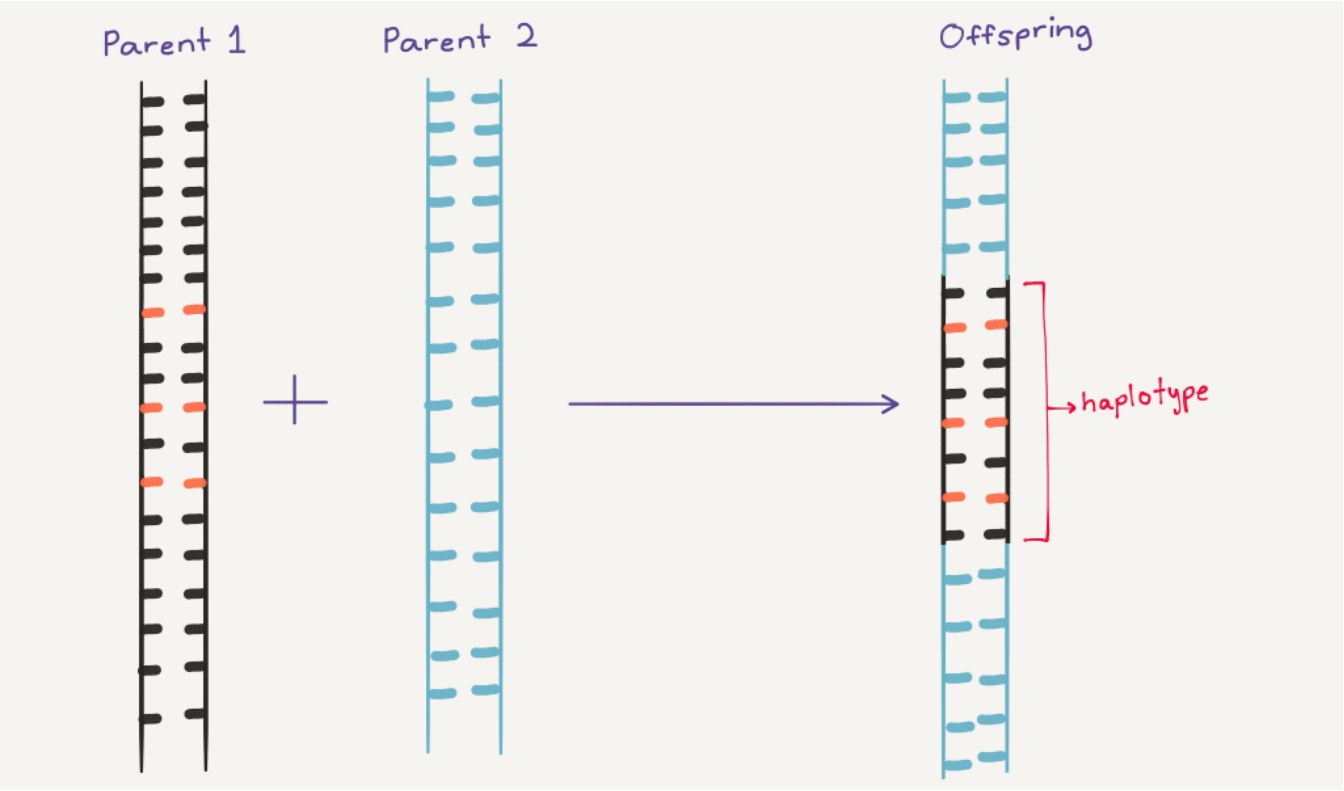
SINGLE NUCLEOTIDE POLYMORPHISM (SNPs)

When a nucleotide is replaced with an incorrect one during DNA replication, the alteration is called a **single nucleotide polymorphism** if it occurs in more than 1% of a population.



Most SNPs occur in non-coding regions of DNA.

If SNPs occur adjacent to each other on the same DNA molecule, they can be inherited together from a single parent. This group of SNPs is called a **haplotype**.



Uses of SNPs

Single nucleotide polymorphisms and **short tandem repeats** are genetic markers.

They can be used as:

- ➔ Indicators of disease susceptibility
    - ◆ Although not necessarily the cause of a disease
- ➔ Determine genetic relatedness
- ➔ Establish evolutionary relatedness

5.5: Inheritance Patterns in a Population

Can population genetic patterns be predicted with any accuracy?

Yes, using DNA analysis and genetic technology.

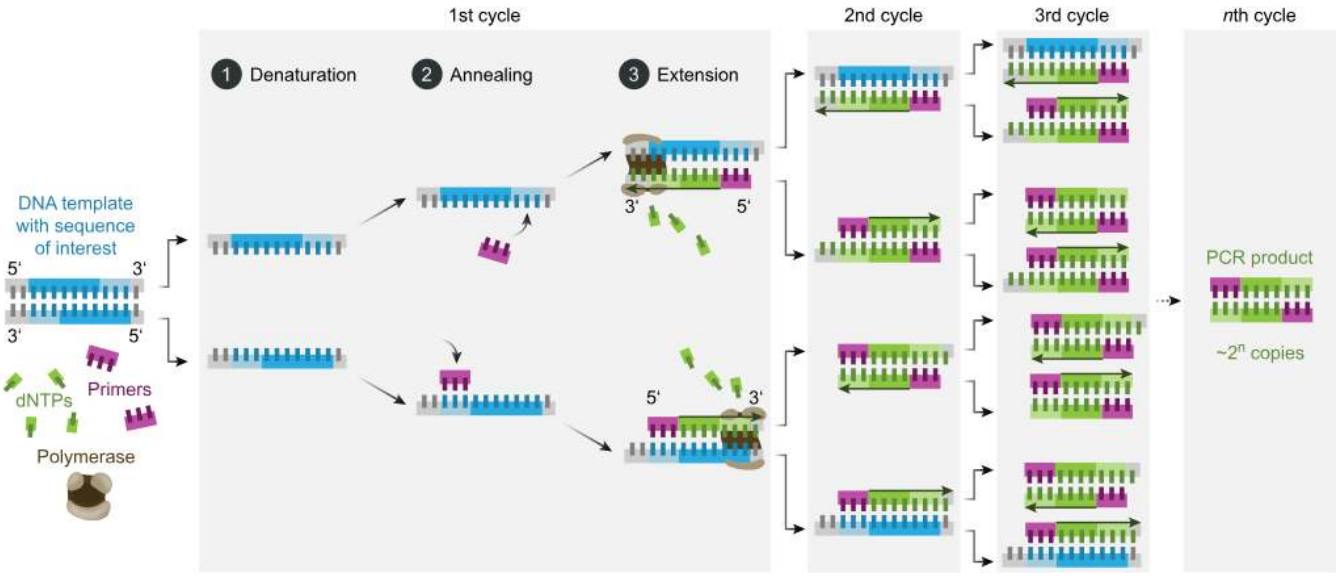
[5.5.1] Technologies Used to Determine Inheritance Patterns

The Human Genome Project

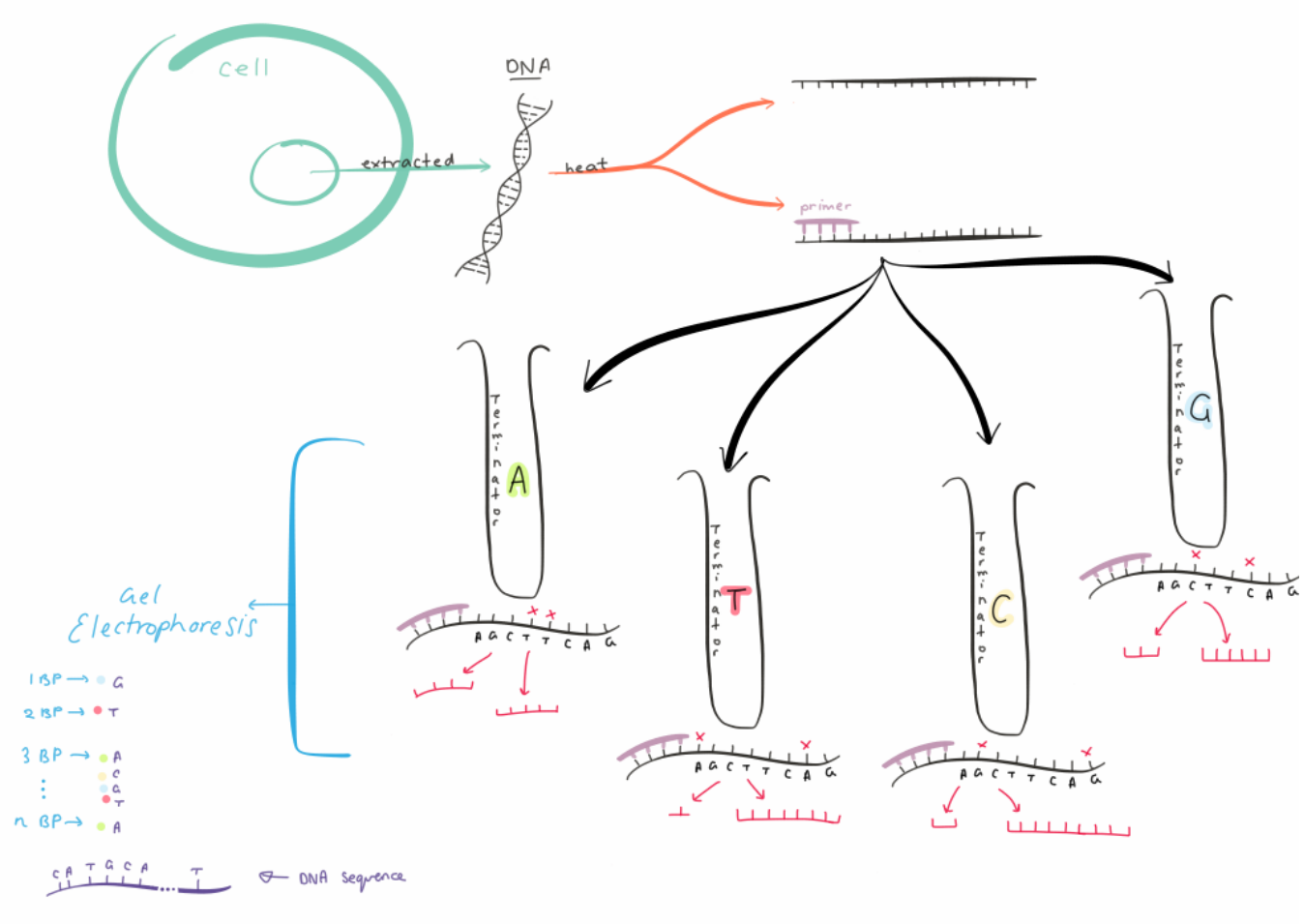
Using DNA sequencing to find the order of nucleotides in every gene of the human genome.

Polymerase chain reaction (PCR)

Isolating	DNA removed from cell.
Denaturing	DNA strands are separated into templates using heat.
Annealing	DNA primers bind to template strand
Extension	DNA polymerase attaches free-floating nucleotides onto template strands



DNA sequencing *(Sanger method)*
Obtains a complete sequence of nucleotides for a specific gene or the whole genome.



1. DNA isolated from cell

2. Strands are separated using heat, so that terminator nucleotides can attach

3. PCR used to amplify DNA of different lengths
    a. Primer initiates beginning of replication
    b. DNA polymerase joins free-floating nucleotides
    c. The replication is stopped by tagged terminator nucleotides

4. The tagged nucleotides will eventually cover each base pair, creating replicated strands of differing lengths (inclusive of final base)

5. Gel electrophoresis is used to find the lengths (in base pairs) of the replicated strands

6. Laser beam used to identify the marker on each length of DNA

7. This shows the terminator nucleotide for each base pair

8. Thus, the order of the base pairs can be found by taking the complement of the terminator nucleotide

DNA profiling
Obtains band patterns using variable regions of DNA (STRs), which can be used to distinguish individuals.

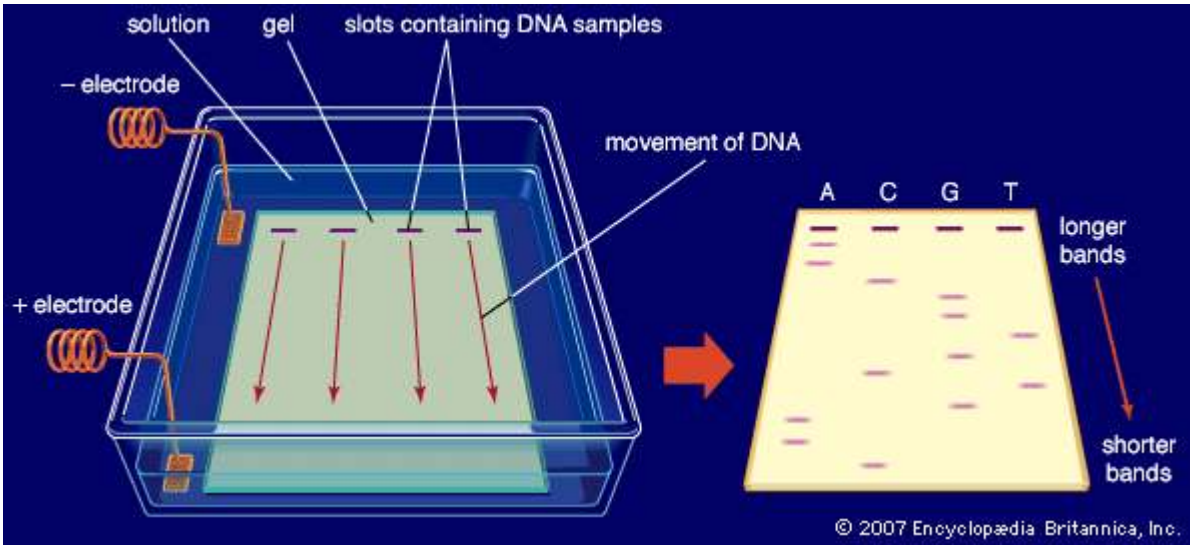
1. DNA sample is extracted

2. DNA is amplified using PCR

3. Restriction endonuclease enzyme cuts out specific short tandem repeats (**STRs**)
   STRs are segments of DNA that are highly variable between individuals.

4. Segments undergo gel electrophoresis

DNA placed into gel wells	→	Electric current passed	→	DNA travels based on size The shorter the segment, the farther it travels	→	Unique band pattern created



5. Radiolabelling probes are added

6. The band pattern is identified

7. Band patterns are unique to individuals (unique DNA profile)

USES OF THE TECHNOLOGIES

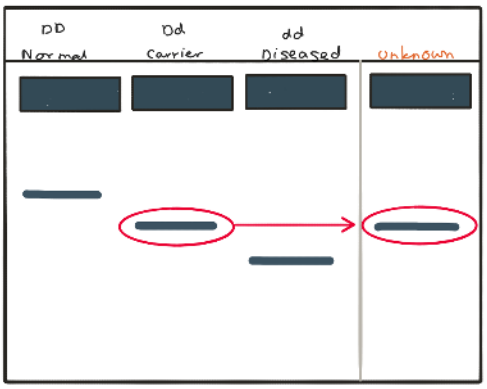
DNA sequencing

- Causes of genetic diseases
- Evolutionary relationships
- Cloning

DNA profiling

Disease testing

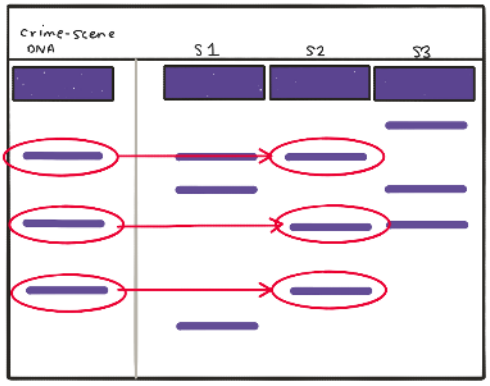
A healthy person, a carrier and a diseased person will have different lengths of DNA sequences. This can be indicated based on how far the DNA moves in electrophoresis.



This individual is most likely a carrier, since the bands match with the known carrier sample.

Forensics

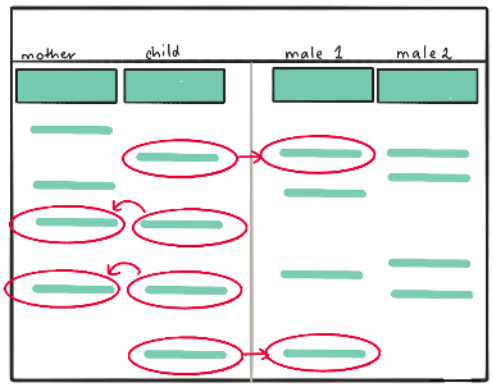
The DNA in crime suspects is compared with DNA from a crime scene.
If it is a 100% match, the suspect is guilty.



The bands of suspect 2 completely match with that from the crime scene, therefore they are probably guilty.

Paternity testing

50% of a child's DNA comes from the mother and the father.
If one parent is known, the other parent is the one which completes the child's DNA sequence.



The bands from male 1 cover the bands not covered by the known mother for the child.
Therefore, he is most likely the father.

[5.5.2] Uses of Population Genetics

CONSERVATION MANAGEMENT

Trends and patterns in population data allow us to:

- ➔ Develop strategies for management of endangered species
- ➔ Observe extinct species as a comparative model for similar endangered animals

The Tasmanian Devil (endangered)

Issues:

- *Virus-induced cheek cancer*
- *Competition from introduced species*
- *Habitat loss*

Management strategies:

- *Vaccines to treat virus*
- *Raised in fenced habitats with low competition*
- *Storage of complete genome to recreate species if they go extinct*

The Wooly Mammoth (extinct)

Observation of DNA from fossils of 2 groups of wooly mammoths:

1. *Mainland group*
2. *Island group*

When compared to the mainland group, the island group had mutations impacting

- *Their sense of smell*
- *Their urine composition*

Which made it difficult for them to mark territory and mate.

This was because there was less gene flow in the island population → less genetic variation.

Thus, the island group lasted much shorter (4300 yrs) than the mainland group (45,000 yrs).

Koalas (endangered)

Tissue samples collected from koalas to identify genetic variation.

Haplotypes studies to identify disease susceptibility.

INHERITANCE OF DISEASE/ DISORDER

Newborns are screened

↓

DNA sequenced

↓

SNPs or haplotype identified

↓

Large-scale database created for whole population

↓

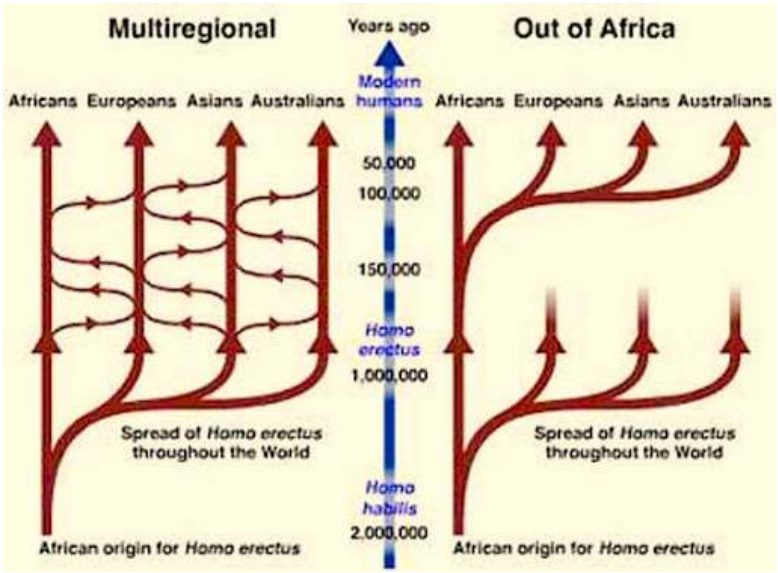
Genomic studies

↓

Develops: - Preventative strategies

- Treatments

HUMAN EVOLUTION

MULTIREGIONAL HYPOTHESIS	REPLACEMENT (OUT-OF-AFRICA) HYPOTHESIS
Process Homoerectus migrated out of Africa ↓ Groups interbred but became reproductively isolated ↓ Different races emerged ↓ Travel prevented complete speciation	**Process** 1st group of ancient humans left Africa ↓ 2nd group evolved into ancestors of the modern human IN Africa ↓ Group 2 outcompeted group 1 ↓ Group 2 spread into different areas and evolved into the modern human ↓ Reproductive isolation led to the races
	
Evidence • Fossils of homosapiens show similarities to homoerectus • Suggests evolutionary relationship	**Evidence** • Carbon dating of fossils suggests ancient and modern humans existed at the same time • Common genes in mt-DNA suggest that they interbred

Using mitochondrial DNA sequencing, we can:

➔ Establish evolutionary history of a species

➔ Establish evolutionary relatedness between species

Evolutionary relatedness of two species can be determined by:

- DNA hybridisation
- % similarity of DNA sequences