Edexcel International AS Biology

Your notes

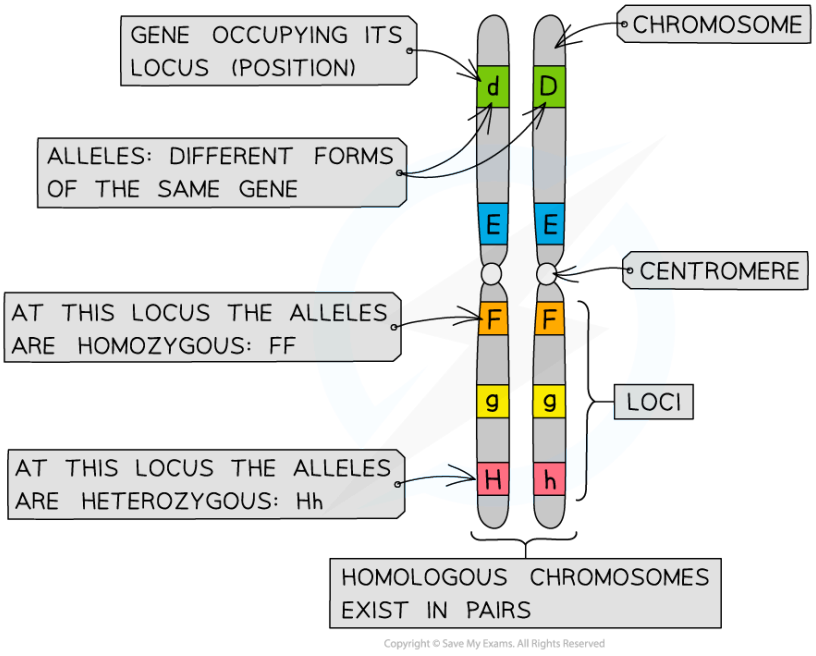
Reproduction & Inheritance

Contents

Your notes

Gene Loci

- Every **chromosome** consists of a long DNA molecule that contains several hundred or even thousands of **different genes** coding for different proteins
  - A length of DNA that codes for a single polypeptide or protein is called a gene
- The position of a gene on a chromosome is known as its **locus** (plural: loci)
  - Through experiments and genetic mapping techniques, scientists have been able to work out the **specific physical locations** of the genes on different chromosomes
  - Each gene occupies a **specific locus** so that the gene for a particular characteristic is always found at the **same position** on a particular chromosome
- Each gene can exist in two or more different forms called **alleles**
- Different alleles of a gene have slightly different nucleotide sequences but they still occupy the **same position** (locus) on the chromosome



Five different genes found at five different loci

Gene Linkage

- Gene loci are said to be **linked** if they are on the **same chromosome**
  - **Loci** (singular: locus) refers to the **specific linear positions** on the chromosome that genes occupy

- Linked genes located on human chromosomes **1 to 22** (i.e. any chromosome that is **not a sex chromosome**, known as **autosomes**) are said to be examples of **autosomal linkage**
- If genes are located on the same **sex chromosome**, they are said to be **sex-linked**

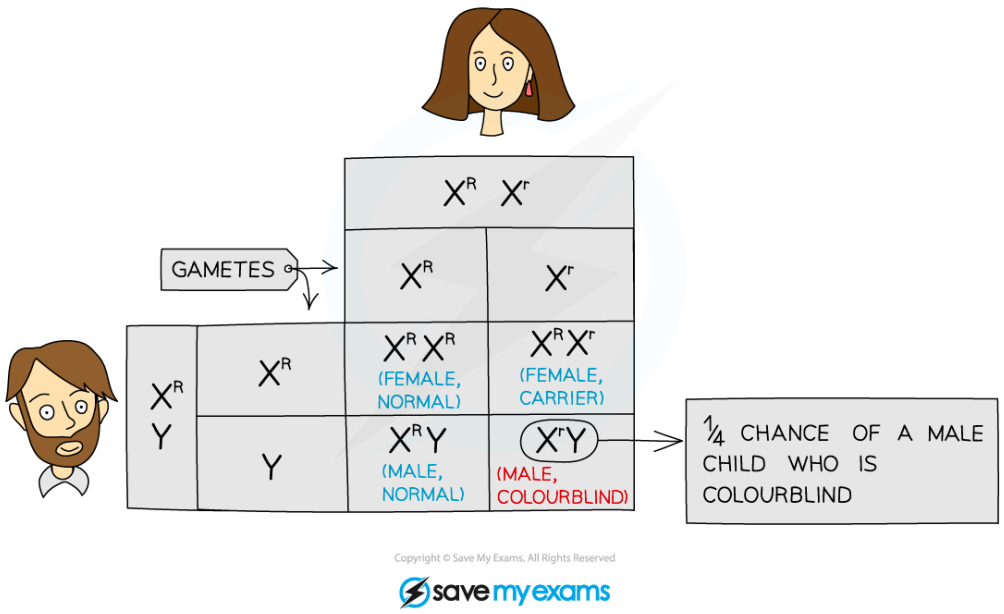
Your notes

Autosomal linkage

- As its name implies, autosomal linkage only occurs on the autosomes (any chromosome that isn't a sex chromosome)
- Two or more genes on the same autosome **do not assort independently during meiosis**
- Instead, these genes are **linked** and they **stay together** in the **original parental combination**
- These linked genes are passed on to offspring **all together** (through the gametes)

Sex linkage

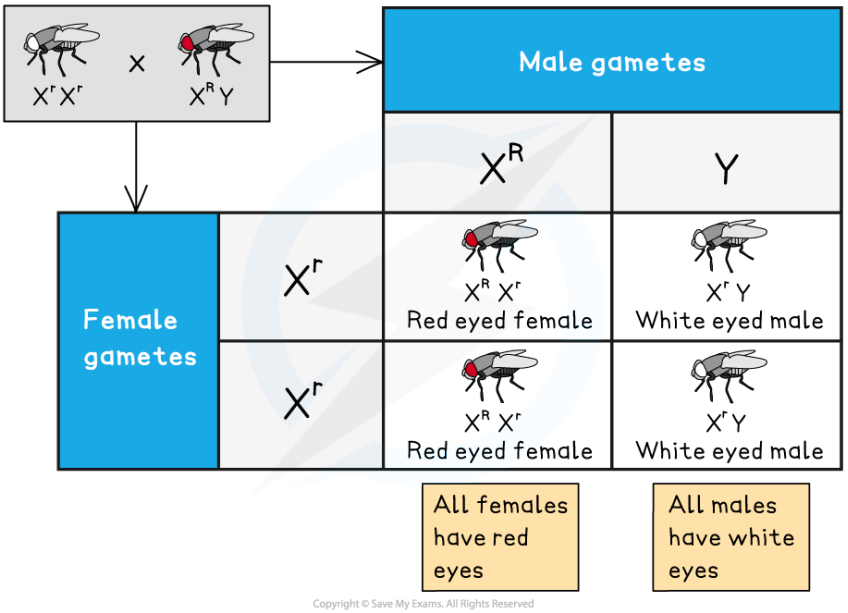
- There are two sex chromosomes: **X** and **Y**
- Females have two copies of the X chromosome (XX), whereas males have one X chromosome and one shorter Y chromosome (XY)
- Some genes are only present on **one sex chromosome** and **not the other**
- As the inheritance of these genes is dependent on the sex of the individual they are known as sex-linked genes
  - Most often sex-linked genes are found on the **longer X chromosome**
- If the gene is on the X chromosome, **males** (XY) will only have **one copy of the gene**, whereas **females** (XX) will have **two**
  - Because males only have one X chromosome, they are much more likely to show **sex-linked recessive conditions** (such as red-green colour blindness and haemophilia)
  - Females, having two copies of the X chromosome, are likely to inherit **one dominant allele** that **masks the effect** of the **recessive allele**
  - A female with one recessive allele masked in this way is known as a **carrier**; she doesn't have the disease, but she has a 50% chance of passing it on to her offspring
  - If that offspring is a male, he will have the disease
- The presence of sex linkage can be **identified** using **pedigree diagrams** and **Punnett squares**
  - When a gene is sex-linked the **phenotypes** are **not spread evenly across the sexes**
  - In the case of a gene that causes a sex-linked disease, one sex will be **disproportionately affected**
  - The results of a cross between a normal male and a female who is a carrier for colour blindness are shown below. In this cross, there is a 25% chance of producing a male who is colourblind, a 25% chance of producing a female carrier, a 25% chance of producing a normal female and a 25% chance of producing a normal male



Punnett square showing the inheritance of colourblindness, an X-linked condition

- Working in the USA in the early 20th century, a scientist called Thomas Hunt Morgan bred fruit flies (*Drosophila melanogaster*) over successive generations
- In his cross-breeding experiments, he came across red-eyed wild types and white-eyed mutants
- He realised there was a **distinct sex bias in the phenotypic distribution** of the **offspring**:
  - All-female offspring of a red-eyed male were red-eyed while all male offspring of a white-eyed female were also white-eyed
- Morgan hypothesised that this occurred because the **gene for eye colour** was located on a **sex chromosome** (i.e. it was **X-linked**)

Your notes



Sex linkage in Drosophila. A cross between a homozygous white-eyed female and a male with red eyes gives all white-eyed males and red-eyed female offspring

Your notes

Your notes

Independent Assortment

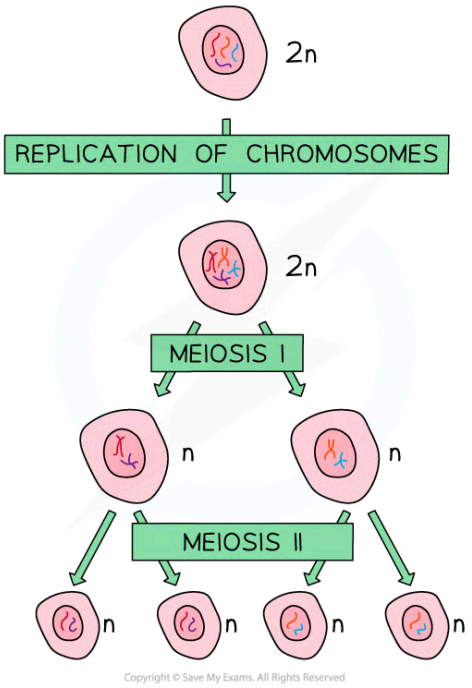
- Meiosis gives rise to cells that are **genetically different** from each other and is the type of cell division used to produce **gametes** (sex cells)
- During meiosis, the nucleus of the original 'parent' cell undergoes **two rounds of division**. These are:
  - **Meiosis I**
  - **Meiosis II**

Meiosis I

- The nucleus of the original 'parent' cell is **diploid** (2n) i.e. it contains **two sets of chromosomes**
- Before meiosis I, these chromosomes **replicate**
- During meiosis I, the homologous pairs of chromosomes are split up, to produce two **haploid** (n) nuclei
  - At this point, each chromosome still consists of **two chromatids**
- Note that the chromosome number halves (from 2n to n) in the **first division** of meiosis (**meiosis I**), not the second division (meiosis II)

Meiosis II

- During meiosis II, the chromatids that make up each chromosome separate to produce **four haploid** (n) nuclei
  - At this point, each chromosome now consists of a **single chromatid**



Your notes

During meiosis, one diploid nucleus divides by meiosis to produce four haploid nuclei

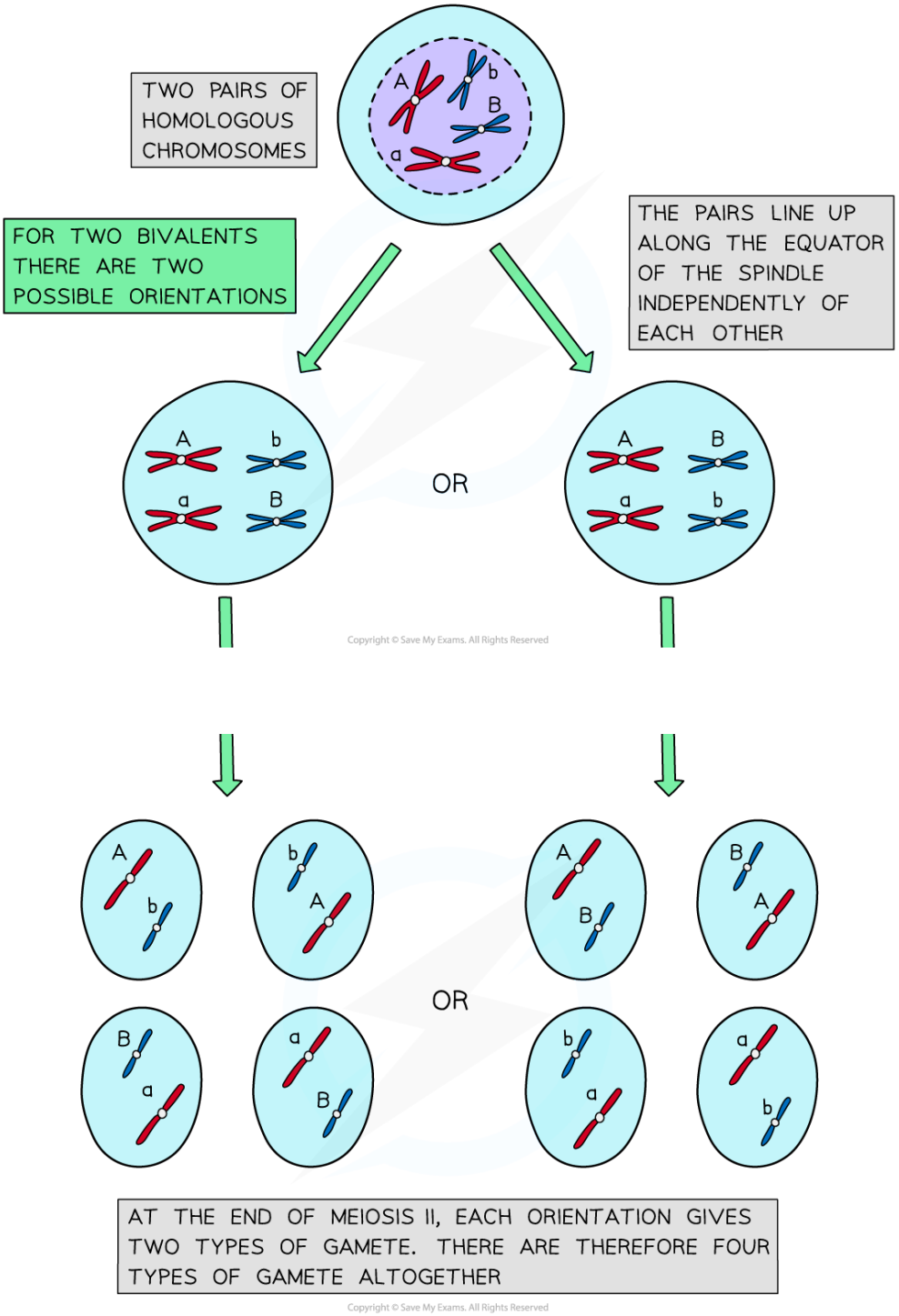
- Having **genetically different offspring** can be **advantageous** for **natural selection**
- Meiosis has several mechanisms that **increase the genetic diversity of gametes** produced. The two main mechanisms are:
  - Independent assortment
  - Crossing over
- Both **independent assortment** and **crossing over** result in **different combinations of alleles** in gametes

Independent assortment

- Independent assortment is the production of **different combinations of alleles** in daughter cells due to the **random alignment of homologous pairs** along the equator of the spindle during **metaphase I** of **meiosis I**
- The different combinations of chromosomes in daughter cells generate an increase in the genetic variation between gametes
- In meiosis I, homologous chromosomes pair up and are pulled towards the equator of the spindle
  - Each pair can be arranged with either chromosome on top, this is **completely random**
  - The orientation of one homologous pair is **independent/unaffected** by the orientation of any other pair
- The homologous chromosomes are then separated and pulled apart to different poles

- The combination of alleles that end up in each daughter cell depends on **how the pairs of homologous chromosomes were lined up**
- To work out the number of different possible chromosome combinations the formula 2^n can be used, where n corresponds to the number of chromosomes in a haploid cell
- For humans, this is 2^{23} which calculates as 8,324,608 different combinations

Your notes

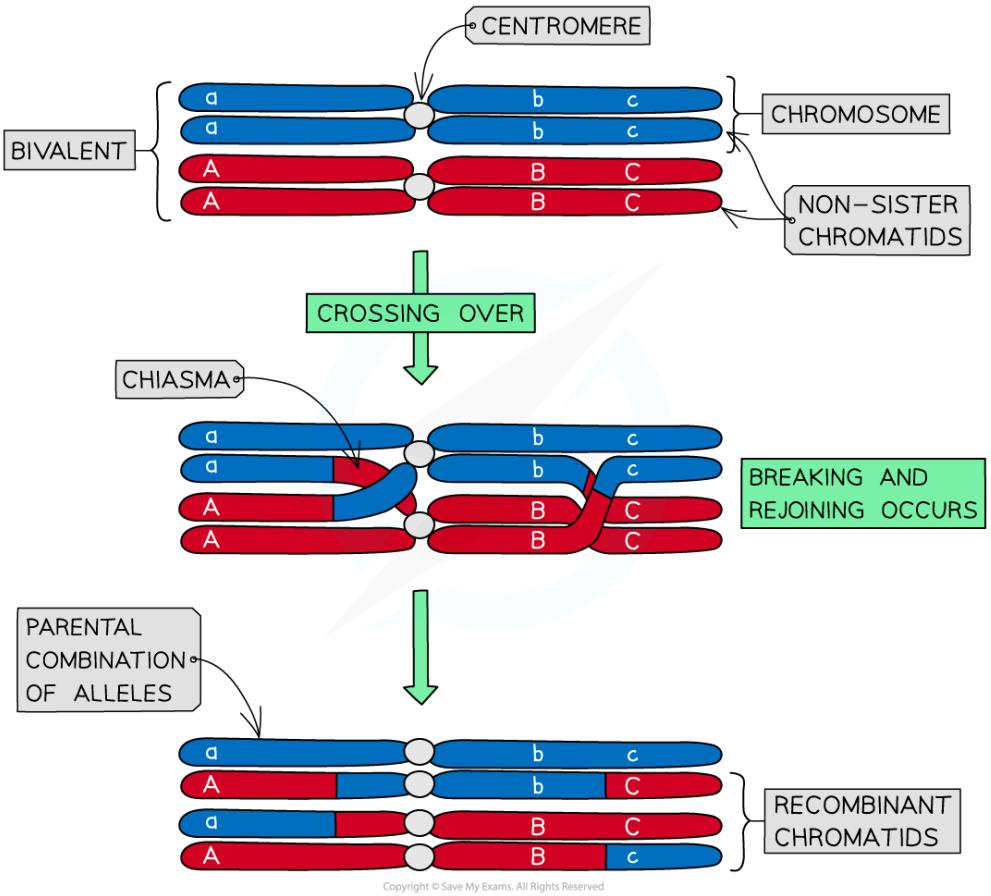


Independent assortment of homologous chromosomes leads to different genetic combinations in daughter cells

Your notes

Crossing Over

- Crossing over is the process by which **non-sister chromatids exchange alleles**
- Process:
  - During **prophase I** of **meiosis I** homologous chromosomes pair up and are in very close proximity to each other
  - The paired chromosomes are known as **bivalents**
  - The non-sister chromatids can **cross over** and get entangled
  - These crossing points are called **chiasmata**
  - The entanglement places stress on the DNA molecules
  - As a result of this, a section of chromatid from one chromosome may **break** and **rejoin** with the chromatid from the **other chromosome**
- This **swapping of alleles** is significant as it can result in a **new combination of alleles on the two chromosomes**
- There is usually at least one, if not more, chiasmata present in each **bivalent** during meiosis
- Crossing over is more likely to occur further down the chromosome away from the centromere



Crossing over of non-sister chromatids leads to the exchange of genetic material

Examiner Tips and Tricks

Several sources of genetic variation have been outlined above. It is also worth remembering that genetic variation can occur on an even smaller scale than chromosomes. **Mutations** can occur within genes. A random mutation that takes place during DNA replication can lead to the production of new alleles and increased genetic variation.

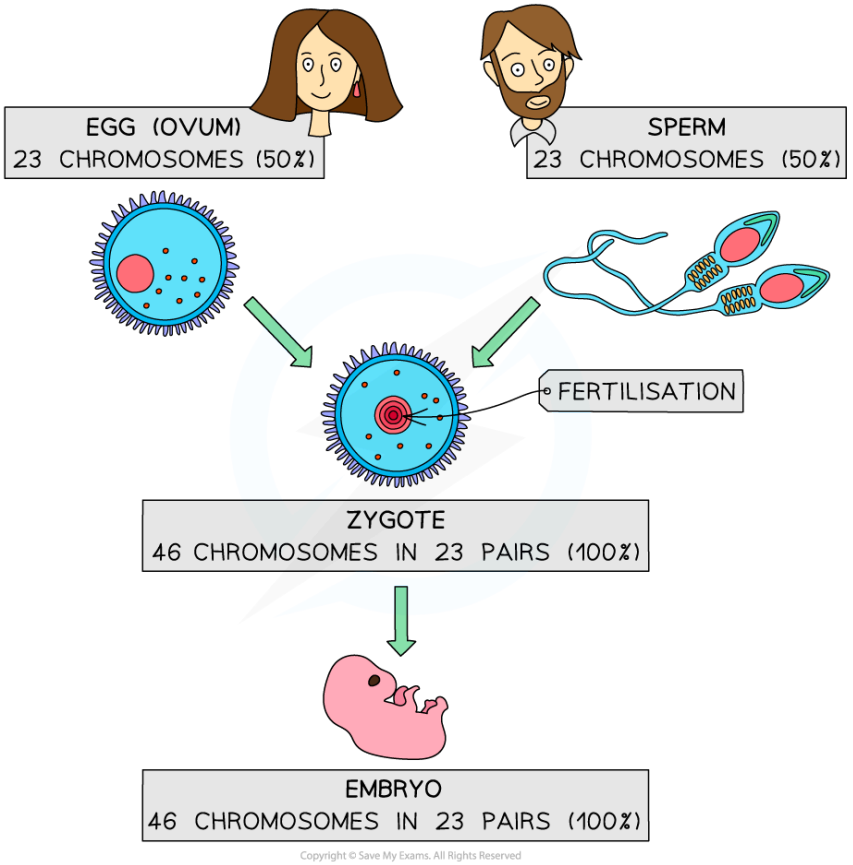
Your notes

Your notes

Gamete Specialisation

Gametes

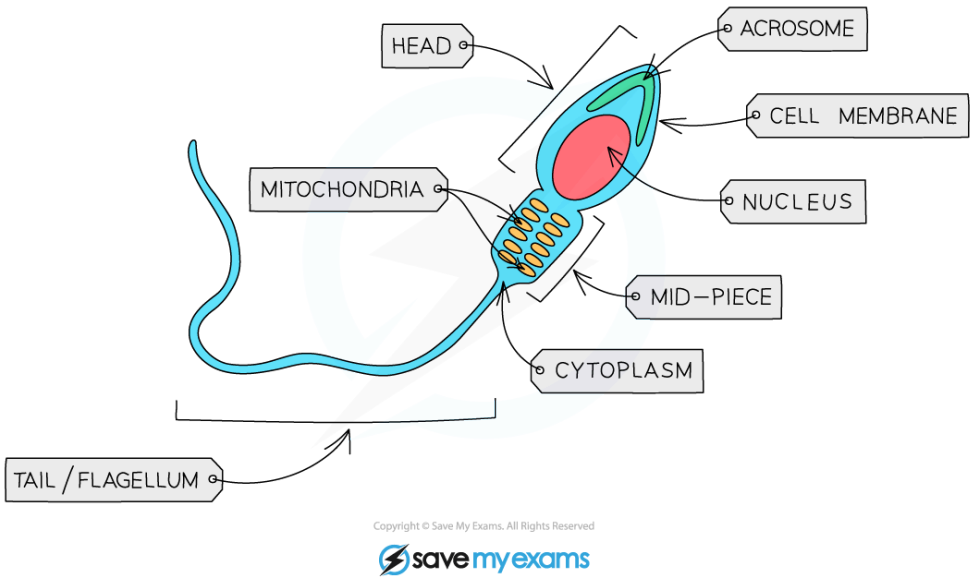
- Gametes are the **sex cells** of an organism
  - For example, the **sperm** and **egg** (ovum) cells in humans
- Gametes fuse during fertilisation to form a **zygote**
  - Fertilisation is the **fusion of the nuclei** from a **male gamete** (sperm cell) and a **female gamete** (egg cell)
- These sex cells are formed during **meiosis** and only have **one copy of each chromosome**, so they are **haploid** cells
  - For humans, that means the sperm and egg cells contain 23 single chromosomes in their nucleus



Sexual reproduction involves the fusing of two gametes to form a zygote that contains DNA from both parents

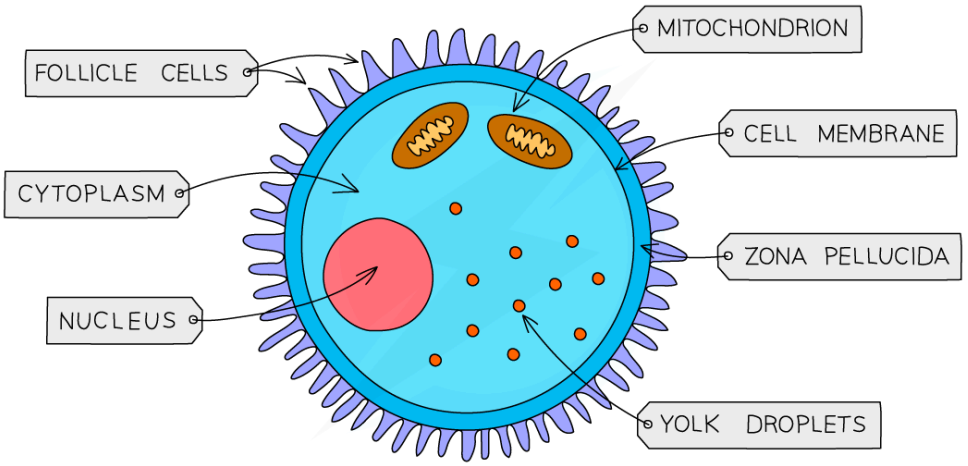
Mammalian gametes are specialised for their functions

- Mammalian gametes have **adaptations** to **increase the chances of fertilisation** and **successful development of an embryo**
- Sperm cells:
  - Have a **flagellum** (tail) that allows them to **swim** towards the egg cell
  - Contain **many mitochondria** that **provide energy for movement** of the flagellum (swimming)
  - An **acrosome** that contains **digestive enzymes** to break down the protective glycoprotein layer (a jelly-like coating known as the **zona pellucida**) surrounding the egg cell - sperm cells must penetrate this layer in order to fertilise the egg
- Egg cells:
  - Are **much larger** than sperm cells as most of their internal space contains **food to nourish a growing embryo**
  - Have follicle cells that form a protective coating
  - Have a jelly-like glycoprotein layer, known as the **zona pellucida**, that **forms an impenetrable barrier after fertilisation** by a sperm cell has occurred, to **prevent other sperm nuclei from entering the egg**
- These features are summarised in the diagrams and tables below



Structure of a mammalian sperm cell

Your notes



Structure of a mammalian egg cell

Adaptations of Mammalian Gametes Table

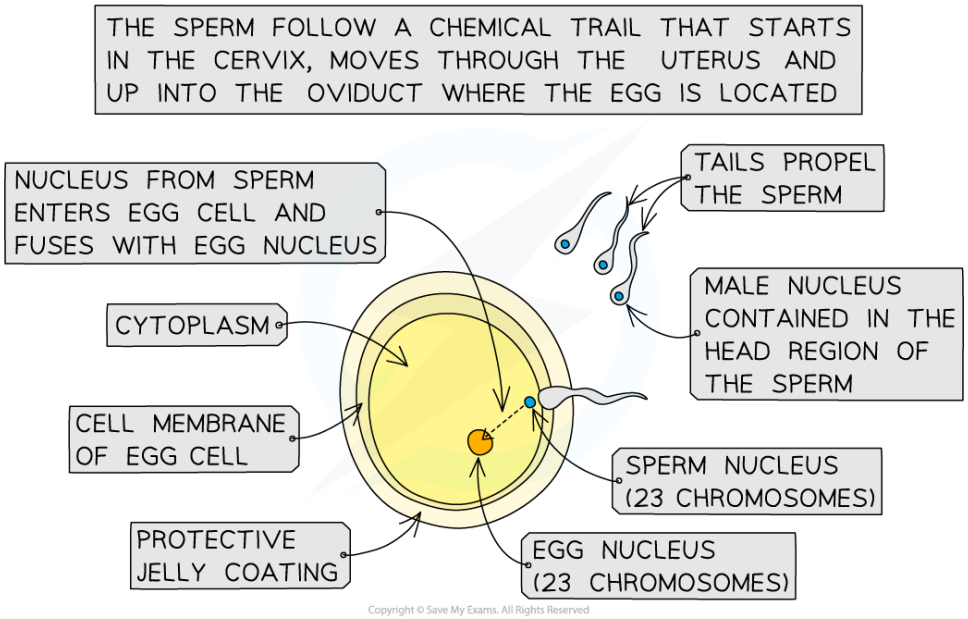
GAMETE	ADAPTIVE FEATURE	REASON
SPERM	HAS A FLAGELLUM (TAIL)	ENABLES IT TO SWIM TO THE EGG
	CONTAINS ENZYMES IN THE HEAD REGION (ACROSOME)	TO DIGEST THROUGH THE JELLY COAT AND CELL MEMBRANE OF AN EGG CELL WHEN IT MEETS ONE
	CONTAINS MANY MITOCHONDRIA	PROVIDE ENERGY FROM RESPIRATION SO THAT THE FLAGELLUM CAN MOVE BACK AND FORTH FOR LOCOMOTION
EGG	CYTOPLASM CONTAINING A STORE OF ENERGY	PROVIDES ENERGY FOR THE DIVIDING ZYGOTE AFTER FERTILISATION
	JELLY LIKE COATING THAT CHANGES AFTER FERTILISATION	FORMS AN IMPENETRABLE BARRIER AFTER FERTILISATION TO PREVENT OTHER SPERM NUCLEI ENTERING THE EGG CELL

Your notes

Your notes

Fertilisation in Mammals

- During sexual intercourse, semen is ejaculated high up into the **vagina** of the female, **near the cervix**
- The sperm cells then follow a **chemical trail released by the egg cell** and travel up through the cervix to reach the **uterus** (the womb)
- The sperm cells then travel into the **oviduct** containing the egg cell
- If a sperm cell meets the egg cell in the oviduct, **fertilisation can occur**
  - This is most likely to occur 1–2 days after the female has ovulated (i.e. released an egg cell from one of her ovaries into an oviduct)
- Fertilisation is the **fusion of the nuclei** from a **male gamete** (sperm cell) and a **female gamete** (egg cell)
  - During fertilisation, the head of a sperm cell releases **enzymes** that **digest** a path through the protective outer layer of the egg cell (the **zona pellucida**), allowing the sperm to pass through the egg cell membrane
    - This process is known as the **acrosome reaction**
  - Once this occurs, the egg cell immediately releases the contents of **vesicles** known as **cortical granules** into the space between the egg cell membrane and the zona pellucida
  - The chemicals contained within the cortical granules cause the **zona pellucida** to **rapidly thicken and harden**, preventing any more sperm cells from entering, **ensuring only one sperm cell can fertilise the egg cell**
    - This process is known as the **cortical reaction**
- The **nucleus** of the sperm cell then enters the egg and fuses with the nucleus of the egg cell
- When the male and female gamete nuclei fuse, they become a **zygote** (fertilised egg cell)
- This zygote contains the full 46 chromosomes (23 pairs of chromosomes), half of which came from the father and half from the mother
- The zygote divides by **mitosis** to form two new cells, which then continue to divide like this until an **embryo** is formed after a few days
- Cell division continues and eventually many of the new cells produced become **specialised** to perform particular functions and form all the body tissues of the offspring



The process of fertilisation in mammals

Your notes

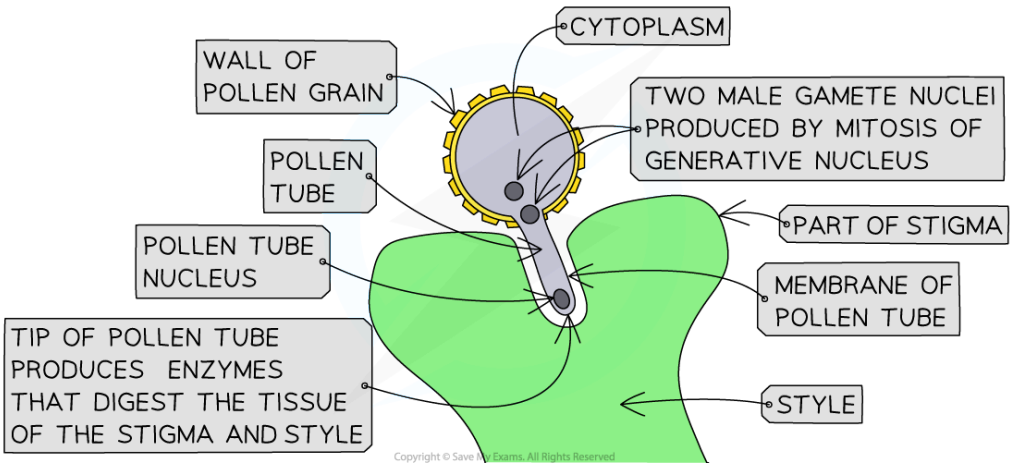
Your notes

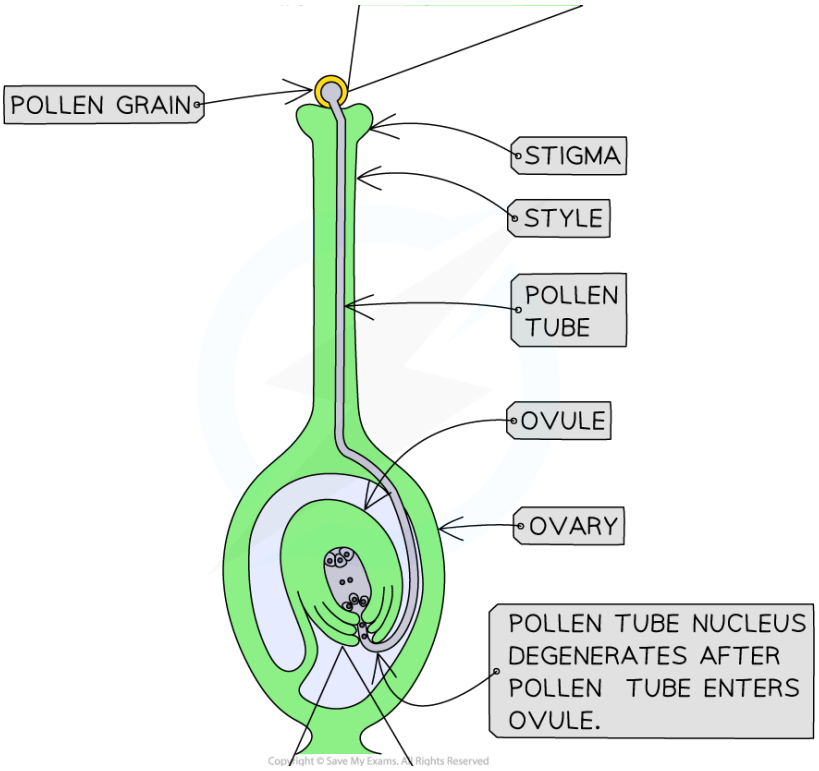
Fertilisation in Flowering Plants

- **Sexual** reproduction in flowering plants requires the **transfer of pollen** between male and female parts of flowers
- The development of flowers occurs in the **reproductive stage** of the plant life cycle
- Flowers contain all the necessary organs and tissues required for **sexual reproduction** by **pollination**
- Key structures of the flower include
  - The **anther** - where pollen is produced
  - The **stigma** - part of the female reproductive organ which receives the pollen
  - The **ovary** - where the female gametes are located
- Flowers usually contain both male and female reproductive parts
- The male reproductive parts produce **pollen**
- The **transferal of pollen from the anther to the stigma** is known as **pollination**

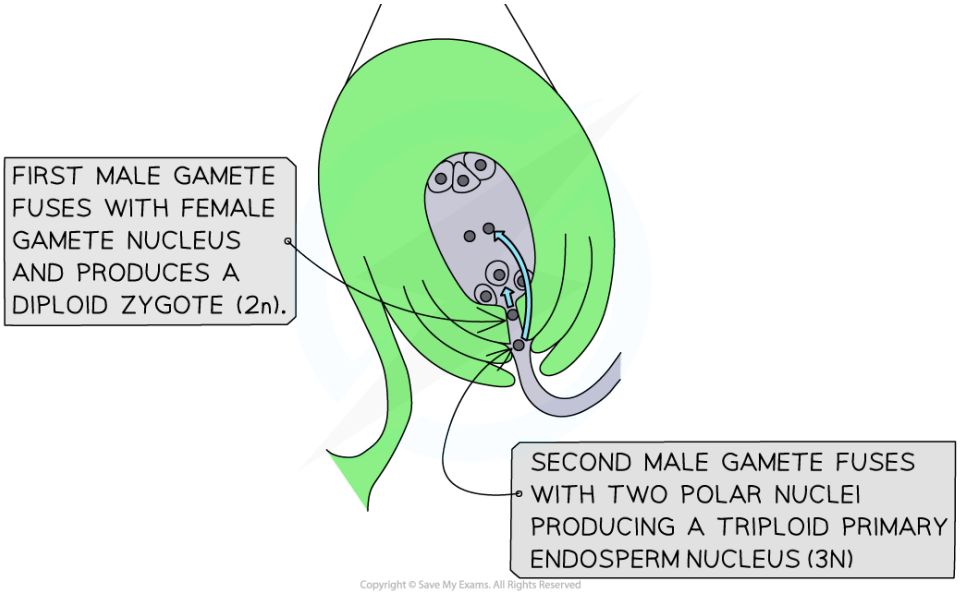
Double fertilisation

- After pollination has occurred, the pollen grain begins to 'grow' or 'germinate' and a **pollen tube** grows from the **pollen grain** down the **style** to the **ovary** of the plant
- As the pollen tube grows towards the ovary, **two haploid male nuclei** move down the tube
  - These are known as the **pollen tube nucleus** and the **generative nucleus**
- At some point, as it travels down the pollen tube, the generative nucleus **divides by mitosis** to form a further **two haploid male nuclei**
  - These are the **male gametes**
  - The two haploid male nuclei travel down the pollen tube towards the female **ovule**
- As the pollen tube reaches the ovule, the **pollen tube nucleus breaks down** and the **two haploid male nuclei** pass **into the ovule** so that **fertilisation** can occur
- In fact, a process known as **double fertilisation** occurs:
  - One haploid male nucleus fuses with the nucleus of the egg cell to form a **diploid zygote**
  - The other haploid male nucleus fuses with **two polar nuclei** present in the ovule to form a **triploid endosperm nucleus**, which will form the **endosperm** (the food supply for the embryo plant when it begins to germinate)


CYTOPLASM
WALL OF POLLEN GRAIN
TWO MALE GAMETE NUCLEI PRODUCED BY MITOSIS OF GENERATIVE NUCLEUS
POLLEN TUBE
PART OF STIGMA
POLLEN TUBE NUCLEUS
MEMBRANE OF POLLEN TUBE
TIP OF POLLEN TUBE PRODUCES ENZYMES THAT DIGEST THE TISSUE OF THE STIGMA AND STYLE
STYLE
Copyright © Save My Exams. All Rights Reserved


POLLEN GRAIN
STIGMA
STYLE
POLLEN TUBE
OVULE
OVARY
POLLEN TUBE NUCLEUS DEGENERATES AFTER POLLEN TUBE ENTERS OVULE.
Copyright © Save My Exams. All Rights Reserved

Your notes



The process of double fertilisation in plants, in which one male nucleus fuses with the two polar nuclei to form the triploid endosperm nucleus and the other fuses with the egg cell to form the diploid zygote

Examiner Tips and Tricks

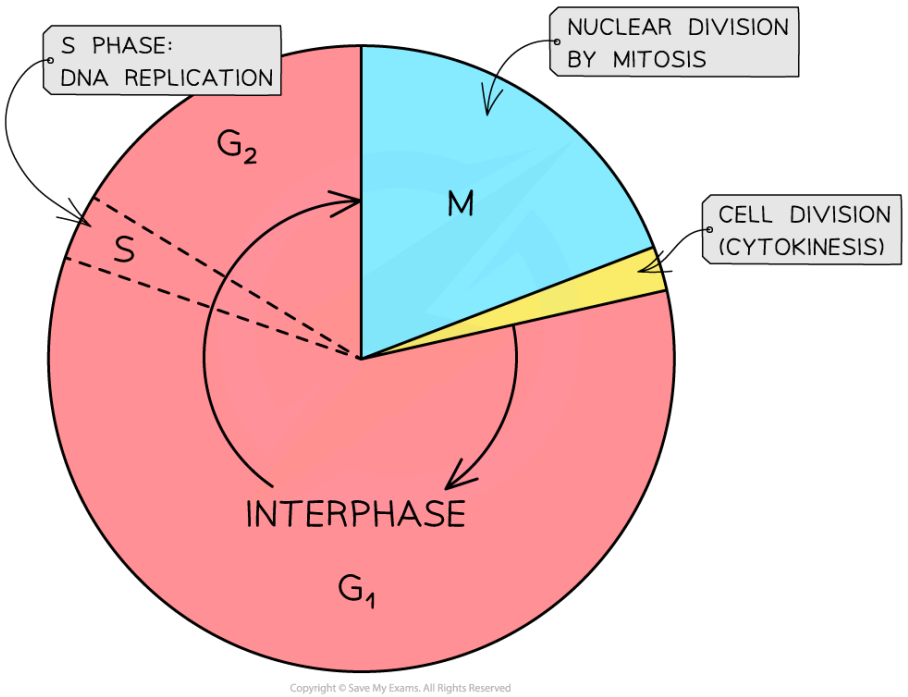
Students often get confused between pollination and fertilisation in plants, but they are not the same thing. Think of pollination as the plant's equivalent to sexual intercourse in mammals – after sexual intercourse, the male sex cells (sperm) have been deposited into the female. But, for fertilisation to occur, the nucleus of a male sperm cell has to fuse with the nucleus of a female sex cell (egg) and the sperm has to travel to find the egg before this happens. It's exactly the same in plants!

Your notes

Your notes

The Cell Cycle

- **Mitosis** is part of a **precisely controlled process** known as the **cell cycle**
- The cell cycle is the **regulated sequence of events** that occurs **between one cell division and the next**
- The cell cycle has three phases:
  - **Interphase**
  - **Nuclear division (mitosis)**
  - **Cell division (cytokinesis)**
- The length of the cell cycle is very variable depending on environmental conditions, the cell type and the organism
  - For example, onion root tip cells divide once every 20 hours (roughly) but human intestine epithelial cells divide once every 10 hours (roughly)
- The movement from one phase to another is triggered by chemical signals called **cyclins**



The stages of the cell cycle

Interphase

- During Interphase the cell **increases in mass and size** and carries out its normal cellular functions, e.g. synthesising proteins and replicating its DNA ready for mitosis

- Interphase consists of **three** phases:
  - **G_1 phase**
  - **S phase**
  - **G_2 phase**
- It is at some point during the **G_1 phase** that a **signal** is received telling the cell to **divide** again
- The **DNA in the nucleus replicates** (resulting in each chromosome consisting of two identical sister chromatids)
  - This phase of the interphase stage of the cell cycle is called the **S phase** – **S** stands for **synthesis** (of DNA)
  - The S phase is relatively short
- The gap between the previous cell division and the S phase is called the **G_1 phase** – **G** stands for **gap**
  - **Cells make the RNA, enzymes and other proteins required for growth**, as well as **replicating organelles** during the G_1 phase
- Between the S phase and next cell division event the **G_2 phase** occurs
  - During the G_2 phase, the **cell continues to grow and the new DNA that has been synthesised is checked** and any errors are usually repaired
  - Other preparations for cell division are made (e.g. production of tubulin protein, which is used to make microtubules for the mitotic spindle)
- **Interphase = G_1 + S + G_2**

Nuclear division (mitosis)

- Follows interphase
- Referred to as the **M phase** – **M** stands for **mitosis**
- **Cell growth stops** during the M phase
- During mitosis, the **two identical sister chromatids** of each chromosome separates from each other and move to opposite poles of the cell
- This ensures that each new nucleus that forms will contain the exact **same genetic information** as the original nucleus

Cytokinesis

- Follows M phase
- Once the nucleus has divided into two genetically identical nuclei, the **whole cell divides** and one nucleus moves into each cell to **create two genetically identical daughter cells**


Your notes

- In animal cells, cytokinesis involves **constriction of the cytoplasm** between the two nuclei and in plant cells a new cell wall is formed

Your notes

Examiner Tips and Tricks

Make sure you know the order of the phases of the cell cycle but also what specifically occurs during the different phases. Don't forget, interphase is itself made up of three distinct stages (G_1, S and G_2) and you need to know what happens during each of these.

For example, an exam question might ask you to identify the stage of the cell cycle during which a cell would be producing the most mRNA molecules and explain why. The correct answer would be the G_1 phase, as this is when protein synthesis is occurring and the production of mRNA occurs during transcription (the first part of protein synthesis).

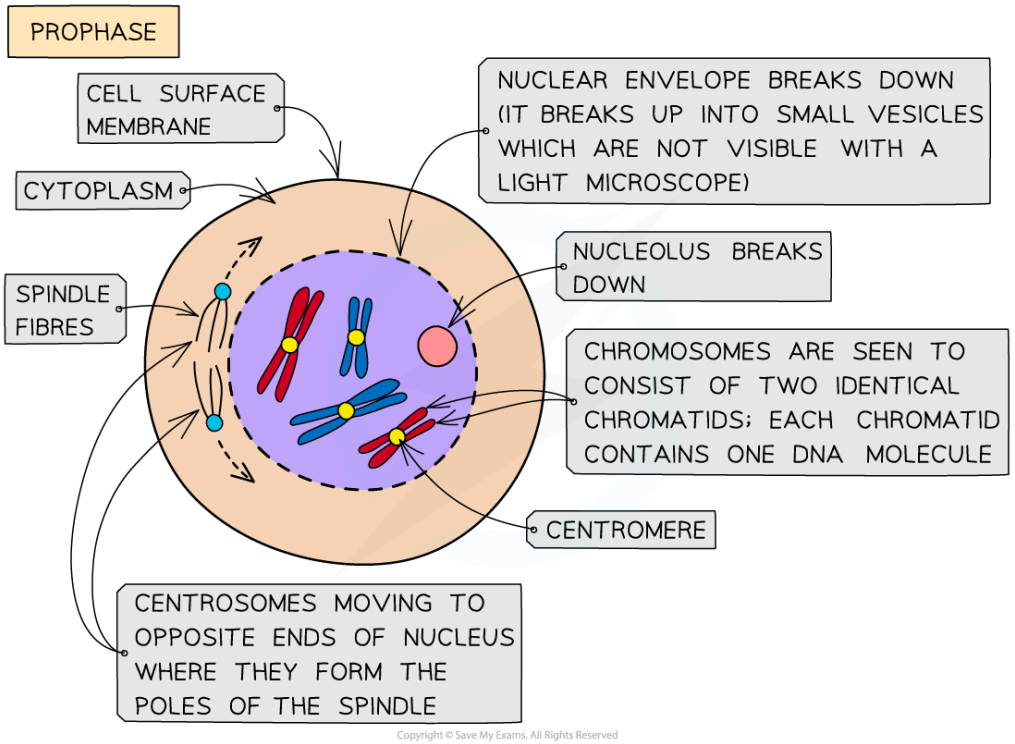
Mitosis

- Mitosis is the process of nuclear division by which **two genetically identical daughter nuclei** are produced that are also genetically identical to the parent cell nucleus (they have the same number of chromosomes as the parent cell)
- Although mitosis is, in reality, one continuous process, it can be divided into **four main stages**
- These stages are:
  - **Prophase**
  - **Metaphase**
  - **Anaphase**
  - **Telophase**
- Most organisms contain many **chromosomes** in the nuclei of their cells (eg. humans have 46) but the diagrams below show mitosis of an animal cell with **only four chromosomes, for the sake of simplicity**
- The different colours of the chromosomes are just to show that half are from the **female parent and half from the male parent**

Prophase

- Chromosomes **condense** and are now visible when stained
- The chromosomes consist of **two identical chromatids** called **sister chromatids** (each containing one DNA molecule) that are joined together at the **centromere**
- The two **centrosomes** (replicated in the G_2 phase just before prophase) move towards **opposite poles** (opposite ends of the nucleus)

- **Spindle fibres** (protein **microtubules**) begin to emerge from the centrosomes (which consist of two **centrioles** in animal cells)
- The **nuclear envelope** (nuclear membrane) **breaks down** into small vesicles

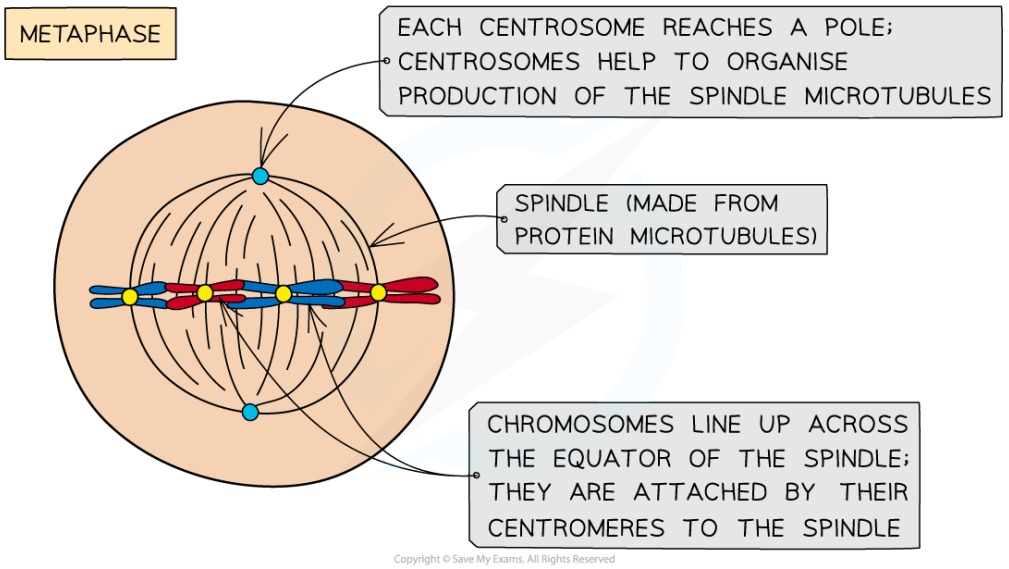


Prophase stage of mitosis where chromosomes condense into visible structures

Metaphase

- **Centrosomes** reach **opposite poles**
- **Spindle fibres** (protein microtubules) continue to **extend from centrosomes**
- Chromosomes **line up at the equator** of the spindle (also known as the metaphase plate) so they are equidistant to the two centrosome poles
- Spindle fibres (protein microtubules) reach the chromosomes and **attach to the centromeres**
- Each **sister chromatid** is attached to a spindle fibre originating from **opposite poles**

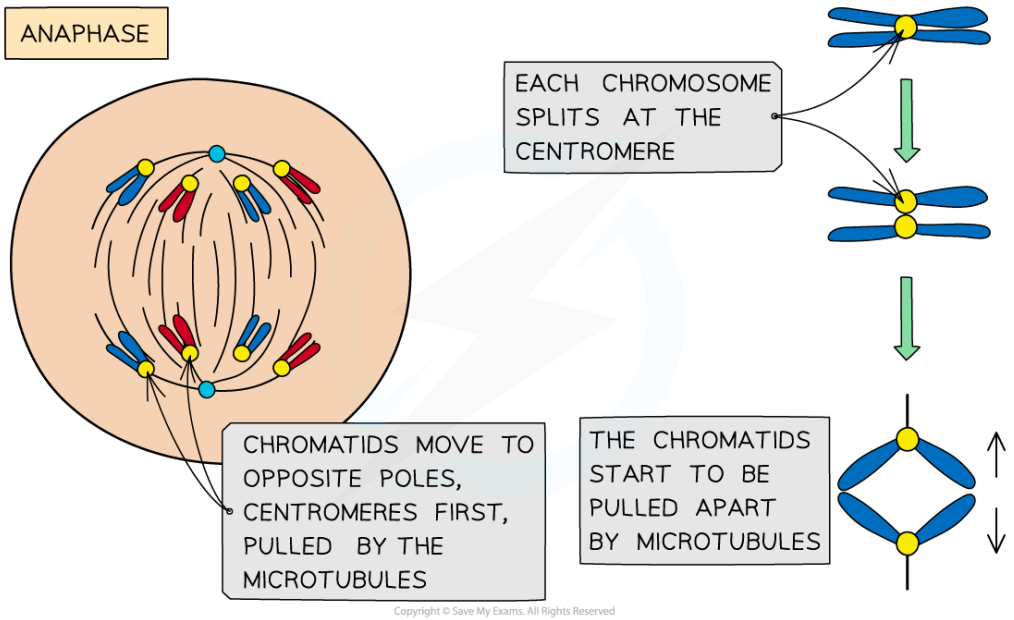
Your notes



Metaphase, where chromosomes line up along the equator of the cell

Anaphase

- The sister chromatids **separate at the centromere** (the centromere divides in two)
- Spindle fibres (protein microtubules) begin to **shorten**
- The separated sister chromatids (**now called chromosomes**) are **pulled to opposite poles** by the spindle fibres (protein microtubules)

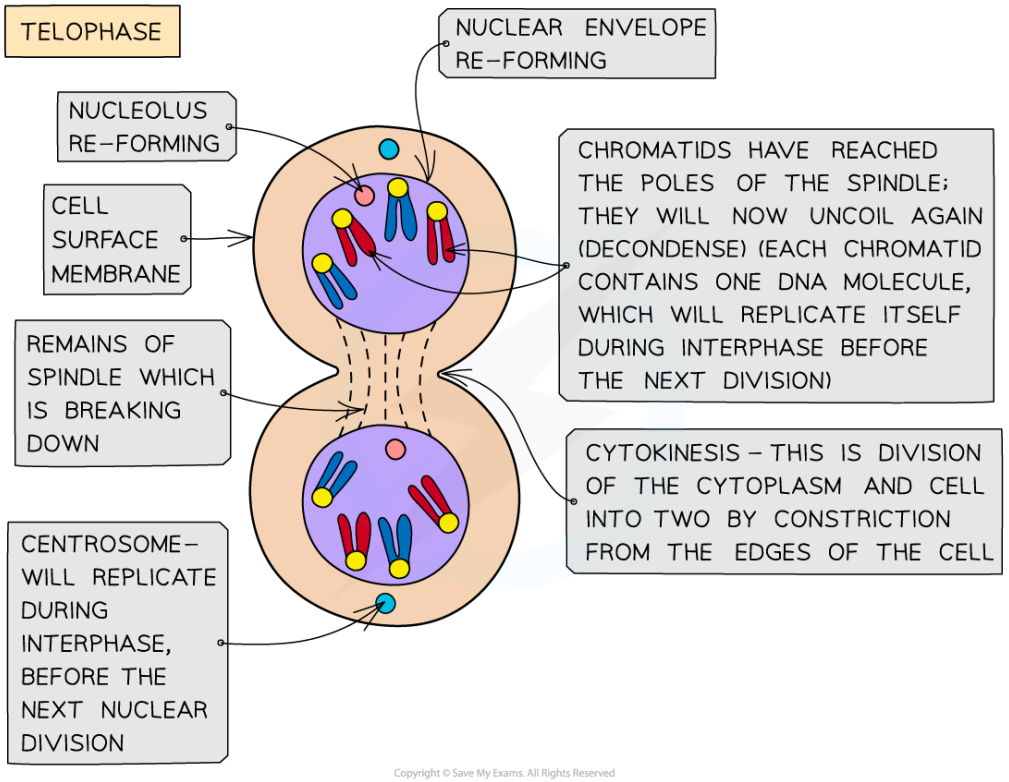


Anaphase, where chromosomes are pulled to the poles of the cell

Telophase

Your notes

- Chromosomes arrive at opposite poles and begin to **decondense**
- **Nuclear envelopes** (nuclear membranes) begin to **reform** around each set of chromosomes
- The **spindle fibres break down**



Telophase, where the nuclei reform and the cell begins to split into two

The significance of mitosis

- The process of mitosis is of great biological significance and is **fundamental to many biological processes**, including:
  - The growth of multicellular organisms
  - The replacement of cells and repair of tissues
  - Asexual reproduction

Growth of multicellular organisms

- The two daughter cells produced are genetically identical to one another (**clones**) and have the same number of chromosomes as the parent cell
- This enables **unicellular zygotes** (as the zygote divides by mitosis) **to grow into multicellular organisms**
- Growth may occur across the whole body of the organism or be confined to certain regions, such as in the meristems (growing points) of plants

Your notes

Replacement of cells & repair of tissues

Your notes

- Damaged tissues can be repaired by mitosis followed by cell division
- As cells are constantly dying they need to be **continually replaced by genetically identical cells**
- In humans, for example, cell replacement occurs particularly rapidly in the skin and the lining of the gut
- Some animals can regenerate body parts, for example, zebrafish can regenerate fins and axolotls regenerate legs and their tail amongst other parts

Asexual reproduction

- Asexual reproduction is the production of new individuals of a species by a single parent organism – the offspring are genetically identical to the parent
- For unicellular organisms such as *Amoeba*, cell division results in the reproduction of a genetically identical offspring
- For multicellular organisms (as seen with many plant species) new individuals grow from the parent organism (by cell division) and then detach ('bud off') from the parent in different ways. Some examples of these are budding in *Hydra* and yeast and runners from strawberries

Examiner Tips and Tricks

Make sure you learn the four stages of mitosis. Cytokinesis is often mistaken as a stage of mitosis, but remember this is a separate part of the cell cycle.

The acronym PMAT can be helpful to remind you what happens during each stage of mitosis:

P = **P**rophase, where the cell **P**repares to divides

M = **M**etaphase, where the chromosomes align along the **M**iddle

A = **A**naphase, where the chromosomes move **A**way from each other

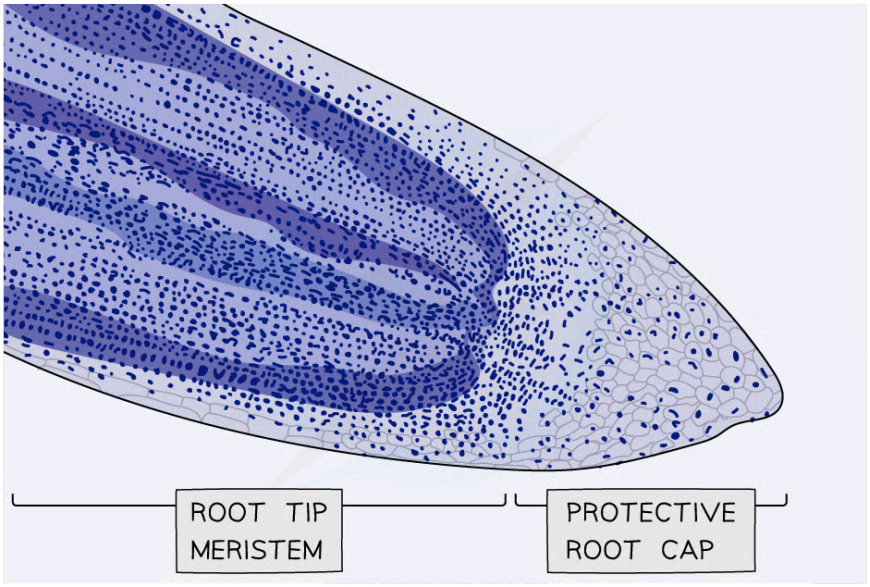
T = **T**elophase, where **T**wo nuclei reform

The chromosome number is important too; after interphase but before the parent cell undergoes mitosis, the human parent cell nucleus actually contains **92 DNA molecules**! This is because during interphase (S phase), the 46 DNA molecules in the parent cell have replicated to form sister chromatids. As human cells have a diploid number of 46 this replication results in 92 molecules. This ensures the two daughter cells will be diploid (have 46 chromosomes each) when mitosis occurs. Remember to read the questions carefully as **only** human diploid cells have 46 chromosomes so if the question refers to another organism, its diploid number will be different.

Your notes

Observing the Stages of Mitosis

- Growth in plants occurs in specific regions called meristems
- The root tip meristem can be used to study **mitosis**
- The root tip meristem can be found **just behind the protective root cap**
- In the root tip meristem, there is a **zone of cell division** that contains cells undergoing **mitosis**
- Pre-prepared slides of root tips can be studied or temporary slides can be prepared using the **squash technique** (root tips are **stained** and then **gently squashed**, spreading the cells out into a thin sheet and allowing individual cells undergoing mitosis to be clearly seen)



A micrograph of a stained root tip, nuclei are clearly visible

Apparatus

- Roots of onion or garlic
- Microscope slide
- Cover slip
- 1 M hydrochloric acid
- Stain (acetic orcein, toluidine blue for example)
- Paper towel

- Scalpel blade
- Cutting tile
- Water bath at 60°C
- Boiling tube
- Optical microscope

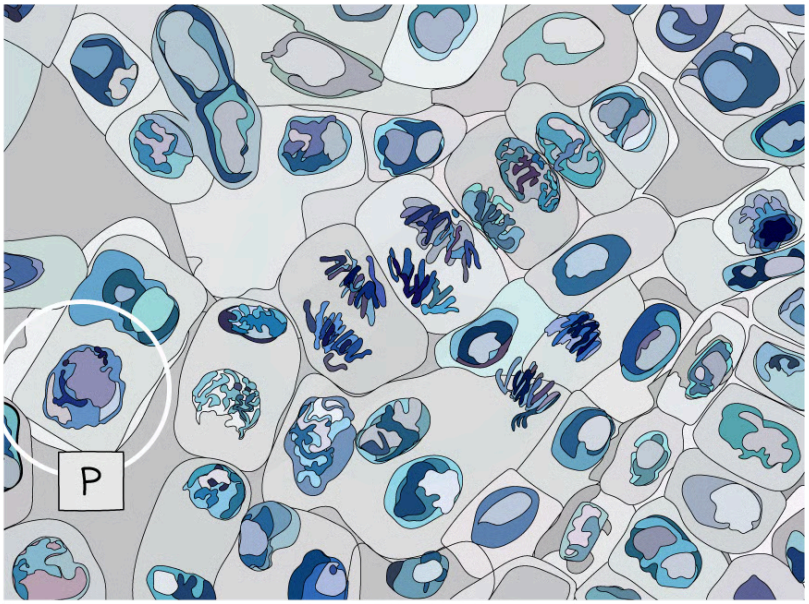
Method

1. **Garlic** or **onion** (*Allium cepa*) root tips are most commonly used (the bulbs can be encouraged to grow roots by suspending them over water for a week or two)
2. Prepare a boiling tube of 1M hydrochloric acid and place in a water bath at 60°C for 10 minutes
3. Remove the **tips** of the roots (about 1cm) and place in the **warmed hydrochloric acid** for 5 minutes
4. Rinse the tips well in cold water using a pipette and blot dry with a paper towel
5. Cut approximately 2mm off the tip and place onto a microscope slide
6. Add a drop of a suitable **stain** (eg. warm, acetic orcein, which stains chromosomes a deep purple)
7. The stained root tip is **gently squashed** on a **glass slide** using a blunt instrument (eg. the handle of a mounting needle or a cover slip using your fist to gently apply pressure in a rolling motion)
8. View the slide under the microscope
9. Cells undergoing mitosis (similar to those in the images below) can be seen and drawn
10. Annotations can then be added to these drawings to show the **different stages of mitosis**

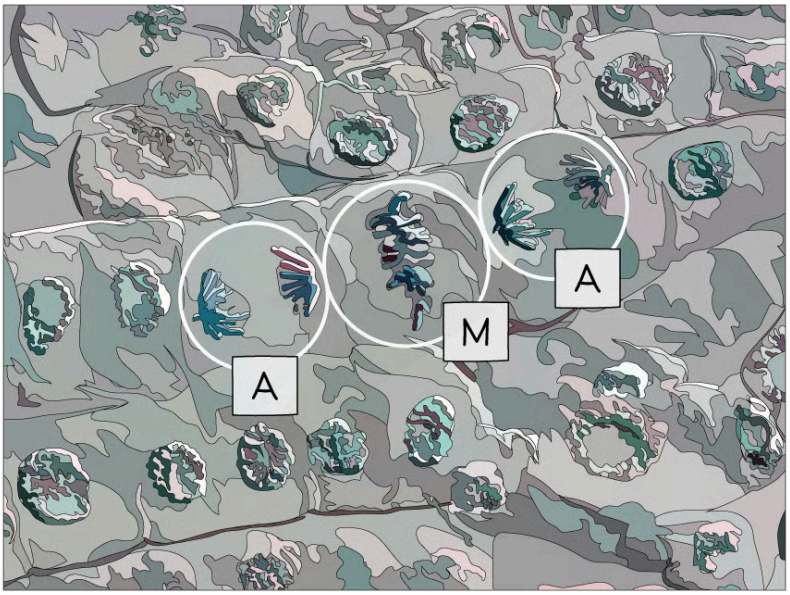
Results

- You should be able to observe cells in different stages of mitosis from the stained meristem

Your notes



Micrograph of cells from a meristem showing cells in prophase (P)



Micrograph of cells undergoing metaphase (M) and anaphase (A)

Your notes

Your notes

Calculation of Mitotic Index

How to calculate mitotic index

- The mitotic index is the **proportion** of cells (in a group of cells or a sample of tissue) that are **undergoing mitosis**
- The mitotic index can be calculated using the formula below:

Mitotic index formula

mitotic index = number of cells with visible chromosomes ÷ total number of cells

- You can multiply the answer by **100** if you need to give the mitotic index as a **percentage**

Worked Example

A student who wanted to observe mitosis prepared a sample of cells. They counted a **total of 42** cells in their sample, **32 of which had visible chromosomes**. Calculate the mitotic index for this sample of cells (give your answer to 2 decimal places).

Answer:

Mitotic index = number of cells with visible chromosomes ÷ total number of cells

- Mitotic index = 32 ÷ 42
- Mitotic index = **0.76**

Worked Example

The table below shows the number of cells in different stages of mitosis in a sample from a garlic root tip. Calculate the mitotic index for this tissue (give your answer to 2 decimal places).

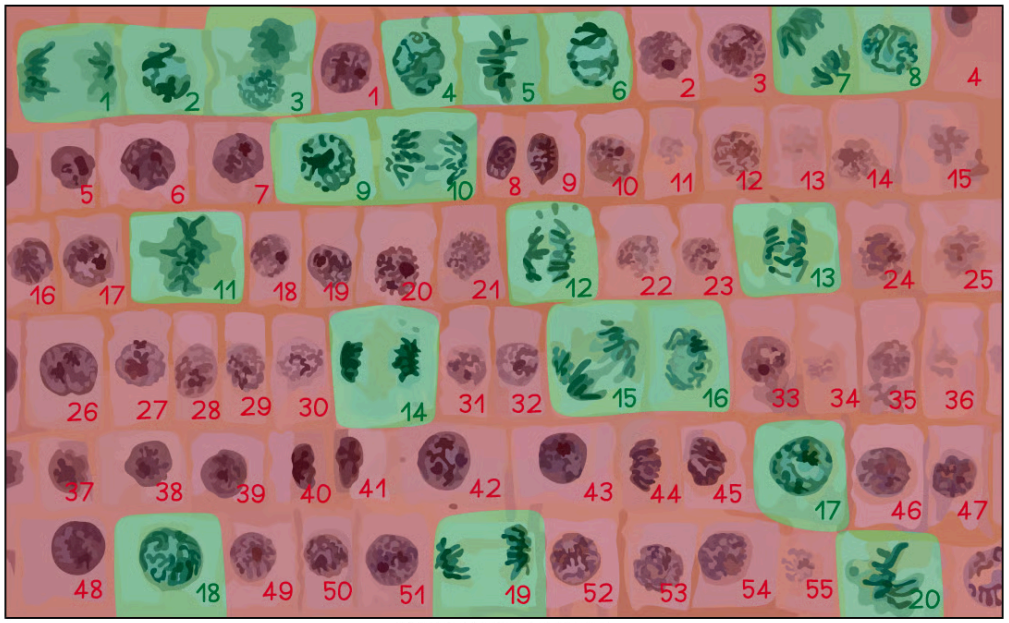
Your notes

Stage of mitosis	Number of cells
Interphase	36
Prophase	14
Metaphase	5
Anaphase	3
Telophase	6

- Mitotic index = number of cells with visible chromosomes ÷ total number of cells
  - Mitotic index = (prophase + metaphase + anaphase + telophase) ÷ total number of cells
  - Mitotic index = (14 + 5 + 3 + 6) ÷ (36 + 14 + 5 + 3 + 6)
  - Mitotic index = 28 ÷ 64
  - Mitotic index = **0.44**

Worked Example

The micrograph below shows a sample of cells from an onion root tip. Calculate the mitotic index for this tissue (give your answer to 2 decimal places).



The cells undergoing mitosis have been identified in green

- Number of cells with visible chromosomes (green) = 20
  - Total number of cells (green + red) = 20 + 55 = 75
  - Mitotic index = number of cells with visible chromosomes ÷ total number of cells
  - Mitotic index = 20 ÷ 75
  - Mitotic index = **0.27**

Your notes

Your notes

Stem Cells

- A **stem cell** is a cell that can divide (by mitosis) an unlimited number of times
- Each new cell (produced when a stem cell divides) has the potential to remain a stem cell or to develop into a **specialised cell** such as a blood cell or a muscle cell (by a process known as **differentiation**)
- This ability of stem cells to differentiate into more specialised cell types is known as **potency**
- There are three main types of potency:
  - **Totipotency** – totipotent stem cells are **embryonic** stem cells that can differentiate into **any cell type found in an embryo**, as well as **extra-embryonic cells** (the cells that make up the **placenta** and **umbilical cord**)
  - **Pluripotency** – pluripotent stem cells are **embryonic** stem cells that can differentiate into any cell type found in an embryo but are **not able** to differentiate into cells forming the placenta and umbilical cord
  - **Multipotency** – multipotent stem cells are **adult** stem cells that have lost some of the potency associated with embryonic stem cells and are **no longer pluripotent**

Totipotent cells

- Totipotent cells can divide and produce **any type of body cell**
- Totipotent cells exist for a **limited time** in early mammalian embryos
- The **zygote** formed when a sperm cell fertilises an egg cell is **totipotent**
- The **embryonic cells up to the 16-cell stage** of human embryo development (around the **fourth day after fertilisation**) are also **totipotent**
  - These cells are still in the form of a solid ball of cells known as a **morula**
- Initially, the totipotent cells in the embryo are **unspecialised**
- During development, totipotent cells begin to translate only part of their DNA, which results in cell specialisation
- There are no totipotent cells present in the later stages of development as cells lose their ability to differentiate into any cell type

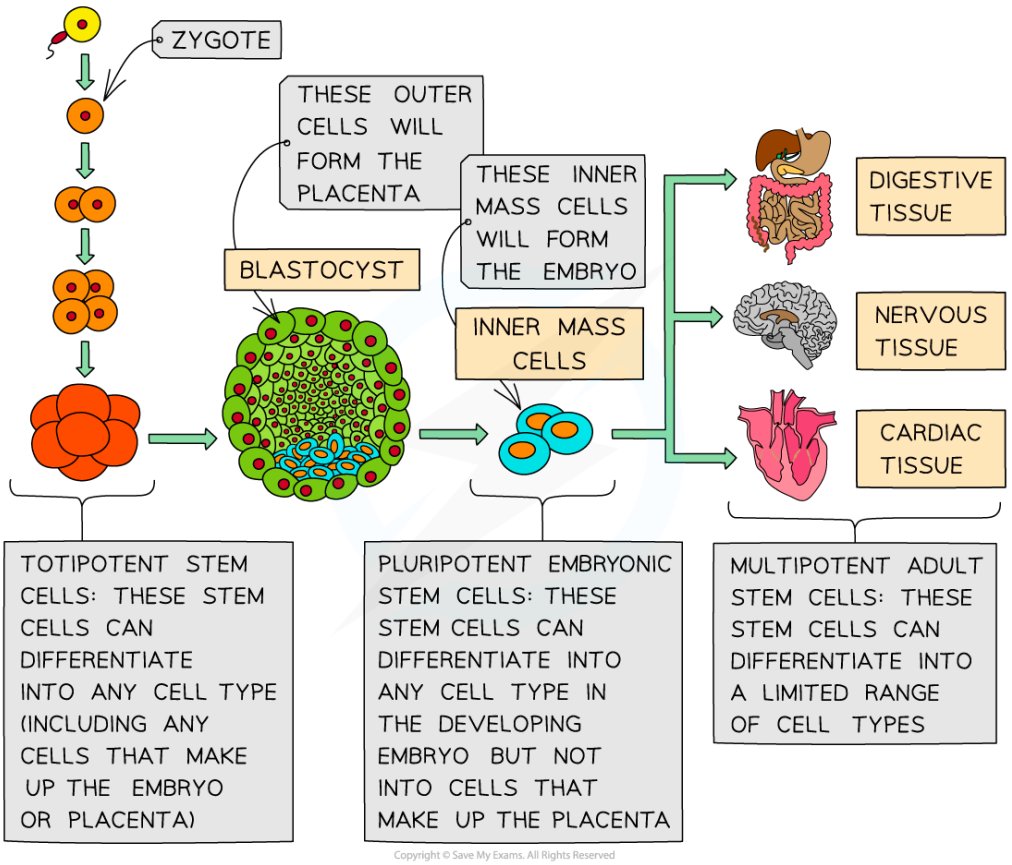
Pluripotent cells

- By around the **fifth day after fertilization**, the embryonic cells have divided further and formed a structure known as a **blastocyst**, which has an **outer layer of cells** and an **inner mass of cells** (that are located inside the outer layer)
- The **outer layer** of cells will later form the **placenta**

- The inner mass cells are **no longer totipotent** (they have lost some of their ability to differentiate)
  - They can still differentiate into any cell type found in an embryo but are **not able** to differentiate into cells forming the placenta and umbilical cord
  - These cells are known as **pluripotent embryonic stem cells**

Multipotent cells

- Stem cells are also found in **some adult tissues** but they are **much less potent** than embryonic stem cells (i.e. they can only specialise into **certain types of cells**)
  - For example, intestinal stem cells specialise into intestinal epithelial cells to replace those that are constantly being lost
- This form of potency is known as **multipotency** – multipotent stem cells are adult stem cells that have lost some of the potency associated with embryonic stem cells and are **no longer pluripotent**
- Plants also contain stem cell in areas of growth, such as their shoots and roots



Stem cells can be totipotent, pluripotent or multipotent

Use of Stem Cells in Medicine

Your notes

- As stem cells have the ability to **differentiate** into other **specialised cell types**, they are very **valuable** in **medical research and treatments** as they have the potential to **replace damaged tissues and cells** (that result from certain diseases)
    - For example, many stem cell therapies already exist, including the treatment for **leukaemia** (bone marrow cancer), in which the existing stem cells in the bone marrow are killed, before being replaced using a **bone marrow stem cell transplant**, which will eventually replace all the bone marrow cells
    - Other stem cell therapies are being researched, including therapies that replace **damaged nerve tissue** to treat **spinal cord injuries** and therapies that replace **damaged heart tissue** to treat **heart disease** and tissue damage caused by **heart attacks**
- There are two sources of human stem cells for use in medicine and research:
    - **Embryonic** stem cells
    - **Adult** stem cells

Your notes

Use of embryonic stem cells

- Due to their ability to differentiate into almost **any cell type**, embryonic stem cells have **huge potential** in the **therapeutic treatment** of many diseases
- For many countries, such as the USA and some countries within the EU, the use of embryonic stem cells is **banned**, even for research
- In other countries, such as the UK, the use of embryonic stem cells is allowed for research but is **very tightly regulated**
- Embryonic stem cells can be one of two potencies:
    - **Totipotent** if taken in the first 3–4 days after fertilisation
    - **Pluripotent** if taken on day 5
- The embryos used for research are often the waste (fertilised) embryos from ***in vitro*** fertilisation (IVF) treatment
    - This means these embryos have the **potential to develop into human beings**
    - This is why many people have **ethical objections** to using them in research or medicine

Use of adult stem cells

- Adult stem cells can divide (by mitosis) an unlimited number of times but they are only able to produce a **limited range of cell types**
- A small number of adult stem cells are found in certain tissues within the body such as:
    - **Bone marrow** - used to produce different types of **blood cell**
    - **Brain** - used to produce different types of **neural and glial cells**
- These small numbers of stem cells remain to produce new cells for the essential processes of growth, cell replacement and tissue repair


Your notes

- Research is being carried out on stem cell therapy, which is the introduction of adult stem cells into damaged tissue to treat diseases (e.g. leukaemia) and injuries (e.g. skin burns)
- The use of adult stem cells is **less controversial** than embryonic stem cells because the donor is able to **give permission**
  - For example, many people donate bone marrow to help treat leukaemia patients
- However, if adult stem cells are being donated from one person to another they need to be a **close match** in terms of **blood type** and other body **antigens**
  - Otherwise, there is a chance that the cells used are **rejected** by the patient's **immune system** (the cells in the stem cell transplant are recognised as being **foreign** and are **attacked** by the patient's immune system)
  - Ideally, the patient's **own** adult stem cells are used to treat them, as there is a **much lower chance of rejection**

Sources of Human Stem Cells Table

Stem Cell	Function	Potential of cell	What can be produced from them
Embryonic stem cell	On the inside layer of an embryo	Undifferentiated / unspecialised	All the different types of specialised cells found in the body
Adult stem cell	Bone marrow	Limited ability to differentiate / partially specialised	Mainly cells of the blood (red blood cells, cells of the immune system)
	Skin	Limited ability to differentiate / partially specialised	Cells found in the different layers of the skin, hair follicles
	Other organs such as the liver and brain	Limited ability to differentiate / partially specialised	Cells found in these organs
	Umbilical cord blood	Limited ability to differentiate / partially specialised	Cells of the blood (red blood cells, white blood cells), muscle and nerve tissue

Evaluating the use of stem cells in medicine

- The use of stem cells that are collected from embryos created via IVF is **ethically questionable** as this results in a **viable embryo** (an embryo that could become a foetus if implanted in a uterus) being **destroyed**

Your notes

- Although their use raises fewer ethical questions, the disadvantage of only using adult stem cells is that, unlike embryonic stem cells, they are unable to differentiate into all of the specialised cell types (some of which may be required to treat certain diseases)
- This means that **society** has to use all the **available scientific knowledge** to make decisions about the use of stem cells (especially embryonic stem cells) in medical therapies, considering all the **arguments for** and **against** their use
- **Official regulatory authorities** are required to help society make these decisions. They do this by comparing the **benefits** and **ethical issues** of stem cell research and making decisions on the extent to which stem cells can be used. These regulatory authorities carry out the following tasks:
  - **Reviewing proposals for scientific research** that uses stem cells and deciding if this research should be allowed to go ahead
  - **Licensing and monitoring of research centres** that are involved in stem cell research
  - **Providing guidelines and codes of practice** for stem cell researchers to ensure they are working to the same high standards
  - **Monitoring developments in scientific research** into stem cell therapies
  - **Providing governments and other professional bodies with correct, up-to-date advice and information** on stem cell research, which in turn helps society to understand how stem cells are being used and why this work is important

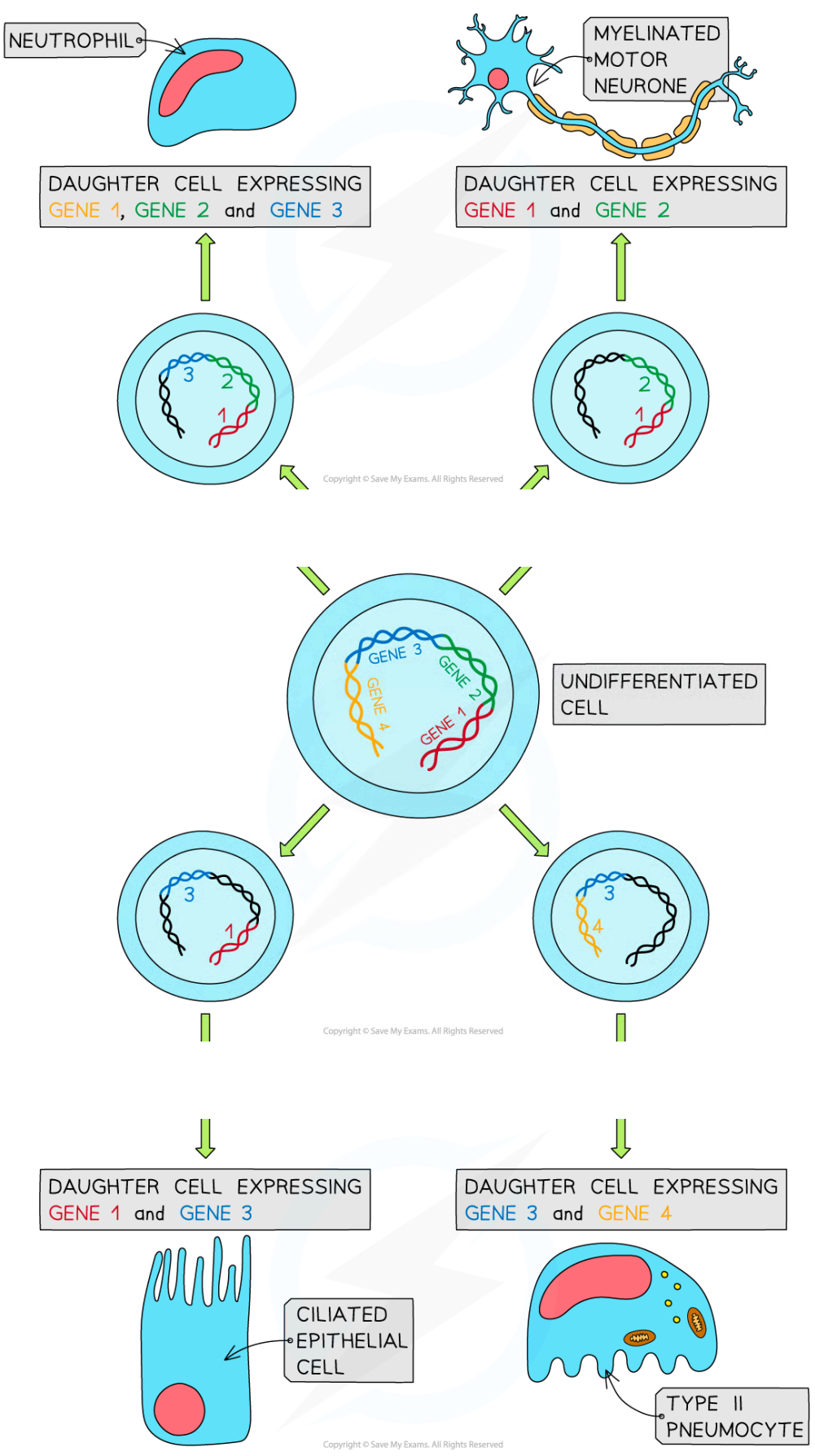
Evaluating the Use of Stem Cells Table

Benefits of using stem cells	Risks / issues of using stem cells	Social issues	Ethical issuess
Great potential to treat a wide-variety of diseases from diabetes and paralysis. Organs developed from a patient's own stem cells reduces the risk of organ rejection and the need to wait for an organ donation. Adult stem cells are already used successfully in a variety of treatments acting as proof of benefits.	Stem cells cultured in the lab could become infected with a virus which could be transmitted to the patient. There is a risk of cultured stem cells accumulating mutations that can lead to them developing into cancer cells. Low numbers of stem cell donors.	It is possible for embryonic stem cells to be collected before birth (from amniotic fluid) or after birth (umbilical cord blood) and stored by a clinic – but this can be expensive and isn't an option for everyone. A lack of peer-reviewed clinical evidence of the success of stem cell treatments. Educating the public sufficiently about what stem cells can and cannot be used for.	Stem cells may be sourced from unused embryos produced in IVF treatment – is it right to use them? Who gives permission? Is it right to create embryos through therapeutic cloning and then destroy them? Who owns the embryo? Should an embryo be treated as a person with human rights? Or as a commodity?

Your notes

Differential Gene Expression

- **Stem cells** become **specialised** through **differential gene expression**
  - This means that only certain genes in the DNA of the stem cell are **activated** and get **expressed**
- Every nucleus within the stem cells of a multicellular organism contains the **same genes**, that is, all stem cells within an organism have an **identical genome**
- Despite the stem cells having the same genome, they are able to specialise into a diverse range of cell types because during **differentiation** certain genes are expressed (**'switched' on**)
- Controlling gene expression is the key to development as stem cells differentiate due to the different genes being expressed
- This differentiation occurs via the following basic steps:
  - Under certain conditions, some genes in a stem cell are **activated**, whilst others are **inactivated**
  - **mRNA** is **transcribed** from **active genes** only
  - This mRNA is then **translated** to form **proteins**
  - These proteins are responsible for **modifying the cell** (e.g. they help to determine the **structure** of the cell and the **processes** that occur within the cell)
  - As these proteins continue to modify the cell, the cell becomes **increasingly specialised**
  - The process of specialisation is **irreversible** (once differentiation has occurred, the cell remains in its specialised form)

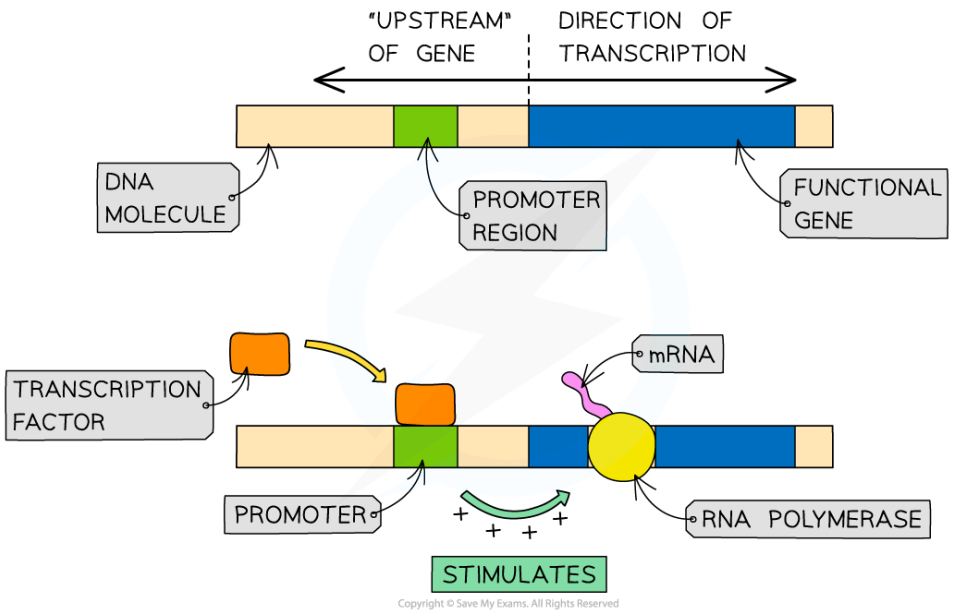

NEUTROPHIL
MYELINATED MOTOR NEURONE
DAUGHTER CELL EXPRESSING GENE 1, GENE 2 and GENE 3
DAUGHTER CELL EXPRESSING GENE 1 and GENE 2
3
2
1
2
1
Copyright © Save My Exams. All Rights Reserved
GENE 3
GENE 2
GENE 4
GENE 1
UNDIFFERENTIATED CELL
3
1
3
4
Copyright © Save My Exams. All Rights Reserved
DAUGHTER CELL EXPRESSING GENE 1 and GENE 3
DAUGHTER CELL EXPRESSING GENE 3 and GENE 4
CILIATED EPITHELIAL CELL
TYPE II PNEUMOCYTE
Copyright © Save My Exams. All Rights Reserved

Your notes

Your notes

Transcription factors control the expression of genes

- Eukaryotes use **transcription factors** to control gene expression
- A transcription factor is a **protein** that controls the transcription of genes by **binding to a specific region of DNA**
- They ensure that genes are being expressed in the correct cells, at the correct time and to the right level
- It is estimated that ~10% of human genes code for transcription factors
  - There are several types of transcription factors that have varying effects on gene expression
  - This is still a relatively young area of research and scientists are working hard to understand how all the different transcription factors function
  - Transcription factors allow organisms to respond to their environment
  - Some hormones achieve their effect via transcription factors
- Transcription factors that **increase** the rate of transcription are known as **activators**
  - Activators work by **helping RNA polymerase to bind** to the DNA at the start of a gene and to begin transcription of that gene
- Transcription factors that **decrease** the rate of transcription are known as **repressors**
  - Repressors work by **stopping RNA polymerase from binding** to the DNA at the start of a gene, inhibiting transcription of that gene
- Some transcription factors bind to the **promoter region** of a gene
  - This binding can either allow or prevent the transcription of the gene from taking place
  - Transcription factors interact with RNA polymerase, either by assisting RNA polymerase binding to the gene (to stimulate expression of the gene) or by preventing it from binding (to inhibit gene expression)
  - Therefore, the presence of a transcription factor will either **increase** or **decrease** the rate of transcription of a gene



In the example above, the transcription factor is an activator as it stimulates the transcription of the gene. Transcription factors, known as repressors, can also inhibit the transcription of genes

Your notes

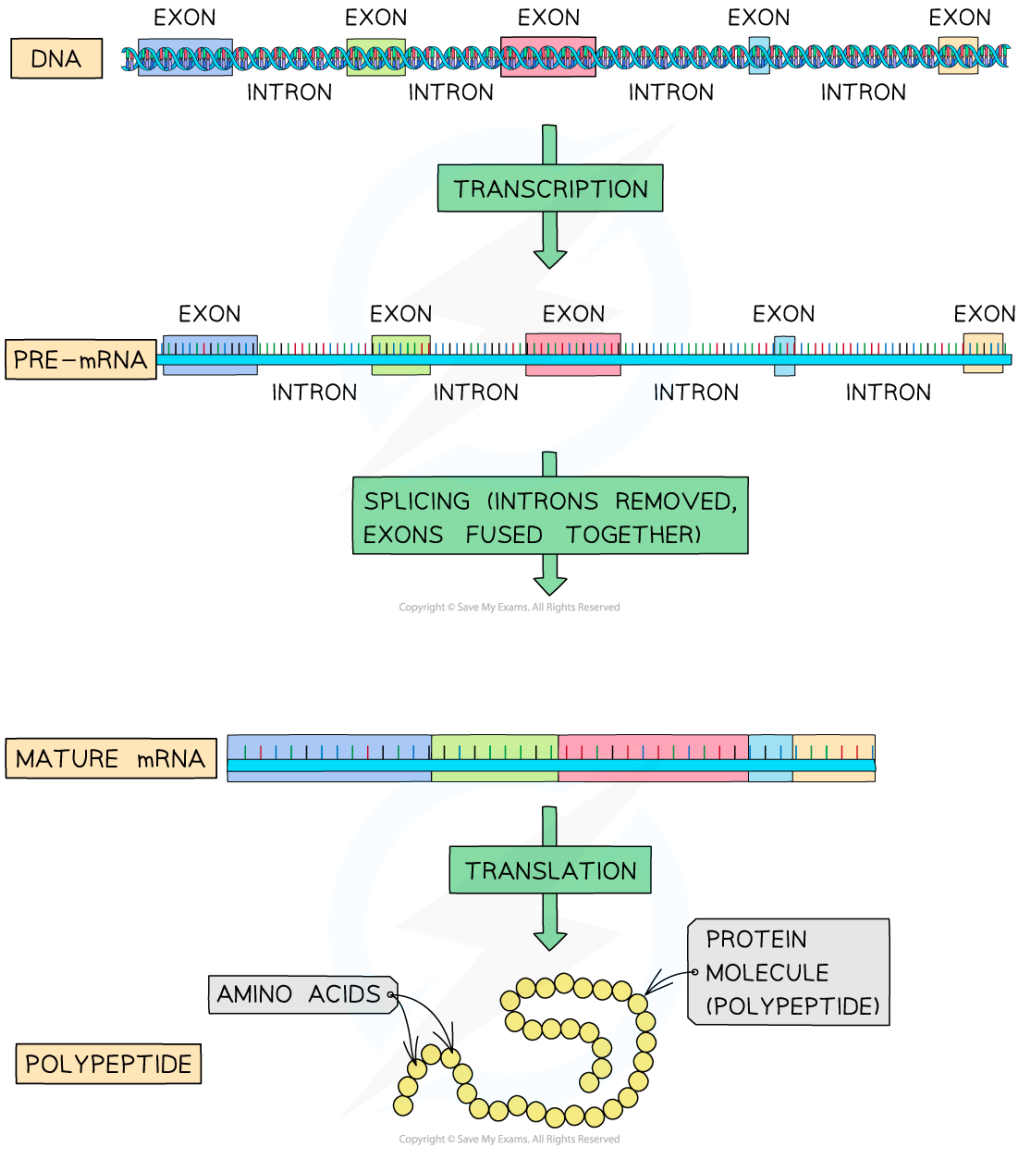
Your notes

One Gene Can Code for More Than One Protein

- **Gene expression** can be **regulated** after an mRNA transcript has been produced
- In eukaryotes, transcription and translation occur in **separate parts of the cell**, allowing for significant **post-transcriptional modification** to occur
- Post-transcriptional modification mechanisms include
  - **Splicing**
  - **Alternative splicing**

Splicing

- Polypeptides are made during the process of protein synthesis, during which the DNA base code is transcribed and translated
- The DNA code within eukaryotic cells contains many **non-coding sections**
- Non-coding DNA can be found within genes; these sections are called **introns**, whilst sections of coding DNA are called **exons**
- During transcription, eukaryotic cells transcribe both introns and exons to produce **pre-mRNA** molecules
- Before the pre-mRNA exits the nucleus, a process called **splicing** occurs
  - The **non-coding intron sections** are **removed**
  - The **coding exon sections** are **joined together**
  - The resulting mRNA molecule contains **only the coding sequences** of the gene
- Since these modifications are made after transcription occurred, they are called **post-transcriptional modifications**

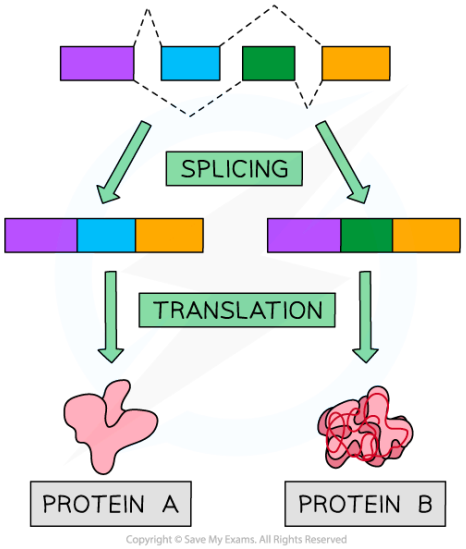


Pre-mRNA is spliced before it exits the nucleus

Alternative splicing

- The exons (coding regions) of genes can be spliced in many different ways to produce **different mature mRNA molecules** through alternative splicing
- A particular exon may or may not be incorporated into the final mature mRNA
- Polypeptides translated from alternatively spliced mRNAs may **differ in their amino acid sequence**, structure and function
- This means that a **single eukaryotic gene can code for multiple proteins**
- This is part of the reason why the **proteome is much bigger than the genome**

Your notes



Alternative splicing of a gene can produce more than one type of protein

Your notes

Examiner Tips and Tricks

It is important you learn the terms pre-mRNA and mRNA, their location and whether they include introns as well as exons. A handy way to distinguish between introns and exons is to remember that **EX**ons are **EX**pressed.


Your notes

Influences on Phenotype

- The **observable characteristics** of an organism are its **phenotype**
- **Phenotypic variation** is the difference in phenotypes between organisms of the same species
- In some cases, phenotypic variation is explained by **genetic factors**
  - For example, the four different blood groups observed in human populations are due to different individuals within the population having two of three possible alleles for the single ABO gene
- In other cases, phenotypic variation is explained by **environmental factors**
  - For example, clones of plants with exactly the same genetic information (DNA) will grow to different heights when grown in **different environmental conditions**
- Phenotypic variation can also be explained by a **combination** of genetic and environmental factors
  - For example, the recessive allele that causes sickle cell anaemia has a high frequency in populations where malaria is prevalent due to heterozygous individuals being resistant to malaria
- The complete phenotype of an organism is determined by the expression of its genotype and the interaction of the environment with this:

Phenotype = Genotype + Environment

Genetic variation

- Organisms of the **same species** will have very **similar genotypes**, but two individuals (even twins) will have differences between their DNA base sequences
- Considering the size of genomes, these differences are small between individuals of the same species
- The small differences in DNA base sequences between individual organisms within a species population is called **genetic variation**
- Genetic variation is transferred from one generation to the next and it generates phenotypic variation within a species population
- Genes can have varying effects on an organism's phenotype:
  - Some characteristics (i.e. the phenotype) are controlled by a **single gene** - these characteristics are known as **monogenic**
    - These characteristics usually show **discontinuous variation** (e.g. blood group)
  - Other characteristics are controlled by **several genes** - these characteristics are known as **polygenic**

- These characteristics usually show **continuous variation** (e.g. height, mass, skin colour)

- Specifically, the different **alleles** an organism has at a **single gene locus** can **determine the phenotype**:
  - Remember - diploid organisms will inherit **two alleles of each gene**, these alleles can be the same or different
  - For example, the F8 gene that codes for the blood-clotting protein Factor VIII. The different alleles at the F8 gene locus dictate whether or not normal Factor VIII is produced and whether the individual has the condition haemophilia

Environmental factors

- The environment that an organism lives in can also have an **impact on its phenotype**
- Different environments around the globe experience very different conditions, including (amongst many factors):
  - Length of sunlight hours (which may be seasonal)
  - Supply of nutrients (food)
  - Availability of water
  - Temperature range
  - Oxygen levels
- Changes in the factors above can affect how organisms **grow and develop**
  - For example, plants with a tall genotype growing in an environment that is **depleted** in minerals, sunlight and water will not be able to grow to their **full potential size determined by genetics** (their phenotype is being affected by their environment)
- Variation in phenotype caused solely by environmental pressures or factors **cannot be inherited** by an organism's offspring (only alterations to the genetic component of gametes will ever be inherited)

Examples of how the environment can affect phenotypic variation

- **Diet** in animals:
  - The fruit fly (Drosophila melanogaster) is **normally grey** but there is a **genetic mutant** that is **yellow** (a genotypic characteristic that is expressed regardless of the environment)
  - If the larvae of normal grey flies are given a diet of silver salts, they develop the yellow colour regardless of their genotype
  - This means that flies that should be grey (according to their **genes**) can become yellow **due to an environmental factor** (their diet)
- **Growing conditions** for plants:


Your notes

- Plants that are grown in the dark or that cannot access enough magnesium become yellow even though, genetically, they should be green
  - This condition is known as chlorosis and occurs because the synthesis of chlorophyll is slowed down or stops completely
- Plants that are grown in the dark may also develop long stems with small, curled leaves even though, genetically, they should develop normally
  - This is known as etiolation

Your notes

Examiner Tips and Tricks

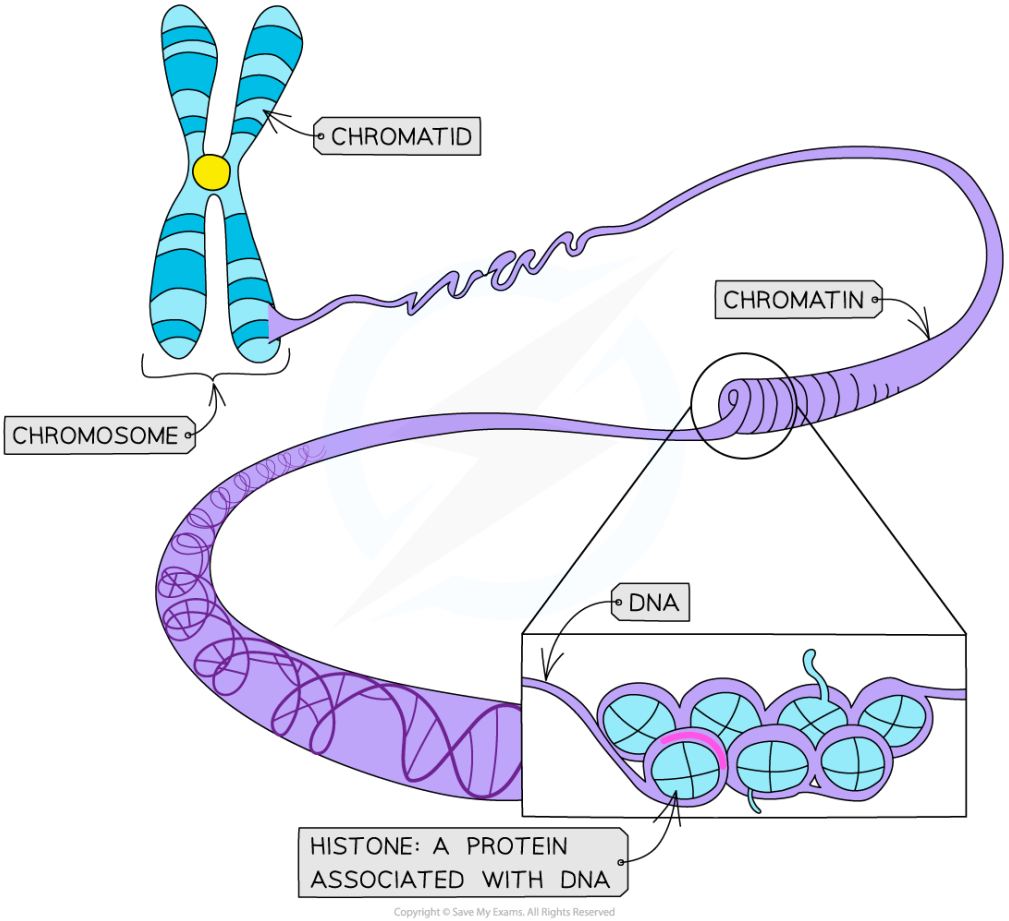
Some questions in the exam may ask you to explain why the variation in phenotype due to genetics is inherited but the variation in phenotype due to environmental factors is not. This is because genetic variation directly affects the DNA of the gametes but variation in phenotype caused by the environment does not.

Epigenetic Modification

- An organism's internal or external **environment** can **influence gene expression** patterns
  - The levels of **regulatory proteins** or **transcription factors** can be affected in response to **environmental stimuli** such as light, and chemicals including drugs and hormones
  - For example, enzymes are activated in response to ultraviolet radiation and increase the expression of melanin-producing genes (and therefore production of melanin), leading to skin pigmentation
- Epigenetics is the control of gene expression by factors other than an individual's DNA sequence
  - Epigenetics involves the **switching-on** and **switching-off** of genes, but **without** changing the actual genetic code
- In eukaryotic cells, nuclear DNA is **wrapped** around **proteins** called **histones** to form **chromatin**
- Chromatin can be **chemically modified** in different ways to alter gene expression, including:
  - **Methylation of DNA** (chemical addition of $-CH_3$ groups)
  - **Histone modification** via **acetylation** of amino acid tails
- Such modifications are called epigenetic tags and collectively, all the epigenetic tags in an organism is called the **epigenome**
- The epigenome can undergo changes due to environmental factors
  - Smoking, stress, exercise and diet can cause epigenetic changes

- Internal signalling from the body's own cells can also cause modifications to occur

- Epigenetic modification is **independent** (i.e. DNA methylation or histone modification in one area is **not linked** to modification in another)
- The chemical modification of histones and DNA controls **how tightly the DNA is wound** around them as the **intermolecular bonding between the histones and DNA changes**
  - If the DNA is wound **more tightly** in a certain area, the genes on this section of DNA are '**switched off**' as the gene and promoter regions are more **hidden from transcription factors and RNA polymerase**
- The modification of histones is **reversible** and therefore can be different in different cell types and can vary with age



DNA is coiled around histone proteins to make chromatin

DNA methylation causes the inactivation of genes

- One method of epigenetic control involves the methylation of DNA
- **Methyl groups** ($-CH_3$) can be **directly added to DNA** to change the activity of a gene
- DNA methylation commonly involves the direct addition of a methyl group to **cytosine bases** which can influence gene expression

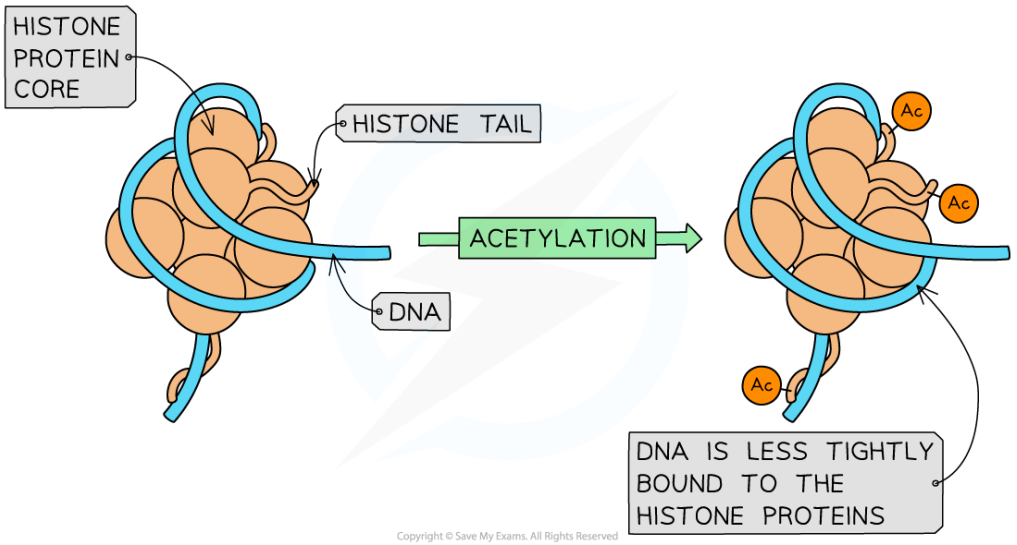
Your notes

- Methylation of DNA **suppresses the transcription of the affected gene** by **inhibiting the binding of transcription factors** and enzymes needed for transcription (e.g. RNA polymerase)
  - Cells use this mechanism to lock genes in the 'off' position
  - The gene is said to be **repressed** or **inactivated**
- DNA methylation can be affected by many environmental, lifestyle or age-related factors

Your notes

Acetylation of histones affects gene expression

- **Acetyl groups** (-$COCH_3$) can be added to lysine amino acids on histone proteins
- Lysine has a **positively charged** R group, this forms ionic bonds with the **negatively charged** phosphate backbone of DNA
  - This helps DNA to coil **tightly** around the histone protein core
- Adding acetyl groups (acetylation) to lysine residues **removes the positive ion** and therefore **removes a bond between the histone protein and the DNA**, this causes the DNA to be **less tightly wrapped**
  - When the DNA is less tightly wrapped, RNA polymerase and transcription factors can **bind more easily** and therefore **gene expression can occur**
  - The gene is said to be **activated**
- Removal of acetyl groups (deacetylation) returns lysine to its positively charged state which has a stronger attraction to the DNA molecule and therefore inhibits transcription and once again stops the gene from being expressed



Acetylation of histones causes chromatin to become less condensed, allowing genes to be transcribed

Epigenetic changes can be passed on following cell division

- Like the genome, the epigenome is **heritable** (i.e. when a cell divides and replicates, epigenetic changes affecting the expression of genes in the DNA of that cell may be **passed on to daughter cells**)
    - For example, during gamete production, DNA in the parent cell usually undergoes de-methylation (the methyl groups are removed) but often methyl groups are not removed and therefore are present in the DNA on the sperm or egg cells
    - One potential explanation for why this occurs is that if an epigenetic change occurs in response to an environmental factor, it may be beneficial for this epigenetic change to also occur in daughter cells (or gametes, for example) so that they are also better adapted for the environmental factor (in the same way the parent cell was)
    - Mounting evidence demonstrates that modifications to the epigenome in one generation can be passed on to the next generation at the cellular or whole organism level

Examiner Tips and Tricks

Epigenetics can be distinguished from mutations, both of which lead to changes in the expression of genes - whilst mutations affect the genetic code itself, by altering nucleotide sequences, epigenetic changes only affect the way the code is read.

Your notes

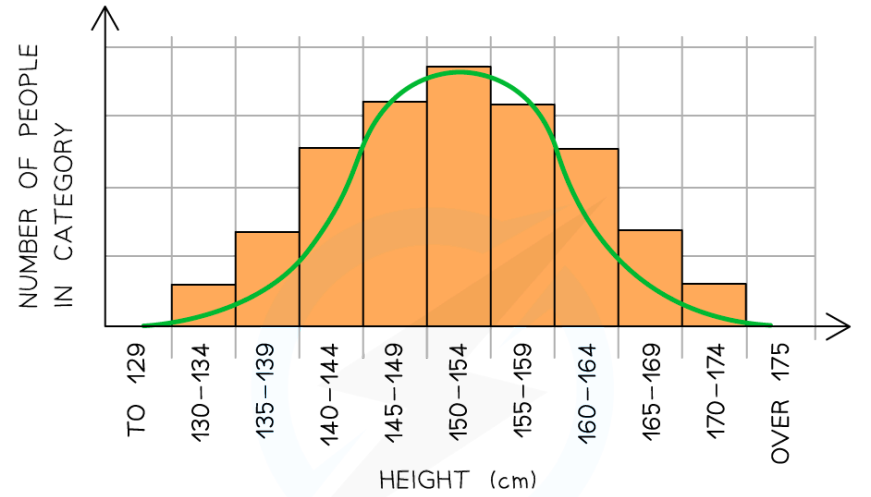
Your notes

Polygenic Inheritance

- Genes can have varying effects on an organism's phenotype:
  - Some characteristics (i.e. the phenotype) are controlled by a **single gene** - these characteristics are known as **monogenic**
    - These characteristics usually show **discontinuous variation** (e.g. blood group)
    - Inheritance of these characteristics is known as **monogenic inheritance**
  - Other characteristics are controlled by **several genes** - these characteristics are known as **polygenic**
    - These characteristics usually show **continuous variation** (e.g. height, mass, skin colour)
    - Inheritance of these characteristics is known as **polygenic inheritance**

Continuous Variation

- Some phenotypes of individuals within a population show **continuous variation**
- The phenotypes **do not fall into discrete categories** (unlike in discontinuous variation)
- Instead, for these features, **a range of values exists between two extremes**, within which the phenotype will fall
  - For example, the mass or height of a human is an example of continuous variation
- The **lack of categories** and the presence of a **range** of values can be used to identify continuous variation when it is presented in a table or graph



FEATURES OF CONTINUOUS VARIATION:

- NO DISTINCT CLASSES OR CATEGORIES EXIST
- CHARACTERISTICS CAN BE MEASURED AND FALL WITHIN A RANGE BETWEEN TWO EXTREMES

Graph showing population variation in height: an example of continuous variation with quantitative differences

Causes of continuous variation

- Some phenotypes are affected by **multiple different genes** or by **multiple alleles** for the same gene at **many different loci** (polygenic inheritance) as well as the environment
  - Phenotype = genotype + environment
- This often gives rise to phenotypes that show **continuous variation**
- At the genetic level:
  - **Different alleles** at a single locus have a **small effect** on the phenotype
  - **Different genes** can have the same effect on the phenotype and these add together to have an **additive effect**
  - If a large number of genes have a **combined effect** on the phenotype they are known as **polygenes**

The additive effect of genes

- The height of a plant is controlled by two unlinked genes **H / h** and **T / t**
- The two genes have an **additive effect**
- The recessive alleles h and t contribute ***x* cm** to the height of the plant
- The dominant alleles H and T contribute **2*x* cm** to the height of the plant

Your notes

- The following genotypes will have the following phenotypes:
  - **h h t t**: $x + x + x + x =$ **4x cm**
  - **H H T T**: $2x + 2x + 2x + 2x =$ **8x cm**
  - **H h T t**: $2x + x + 2x + x =$ **6x cm**
  - **H H T t**: $2x + 2x + 2x + x =$ **7x cm**
  - **H h T T**: $2x + x + 2x + 2x =$ **7x cm**
  - **h h T t**: $x + x + 2x + x =$ **5x cm**
  - **H h t t**: $2x + x + x + x =$ **5x cm**

Examiner Tips and Tricks

Be careful when answering questions that involve polygenes or genes with an additive effect. It is not a given that each gene will have the same effect on the phenotype as in the example above so make sure to double check the information you have been given in the question.

Your notes