DP IB Biology: HL

3.2 Meiosis

Contents

Your notes

3.2.1 Meiosis

Meiosis

- There are two processes by which the **nucleus** of a **eukaryotic cell** can **divide**. These are:
    - Mitosis
    - Meiosis

- **Mitosis** gives rise to **genetically identical cells** and is the type of cell division used for **growth**, **repair** of damaged tissues, **replacement** of cells and **asexual reproduction**
- **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:
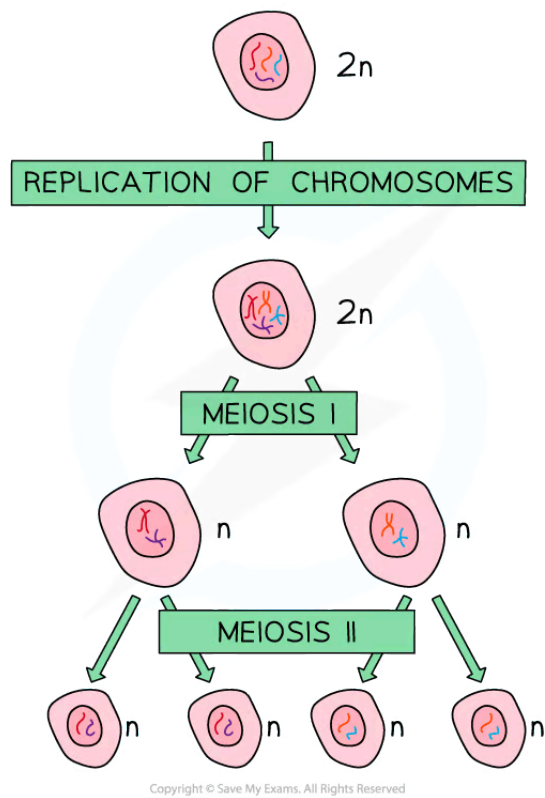   - **Meiosis I**
    - **Meiosis II**

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**

One diploid nucleus divides by meiosis to produce four haploid nuclei

Discovery of Meiosis

NOS: Making careful observations; meiosis was discovered by microscope examinations of dividing germ-line cells
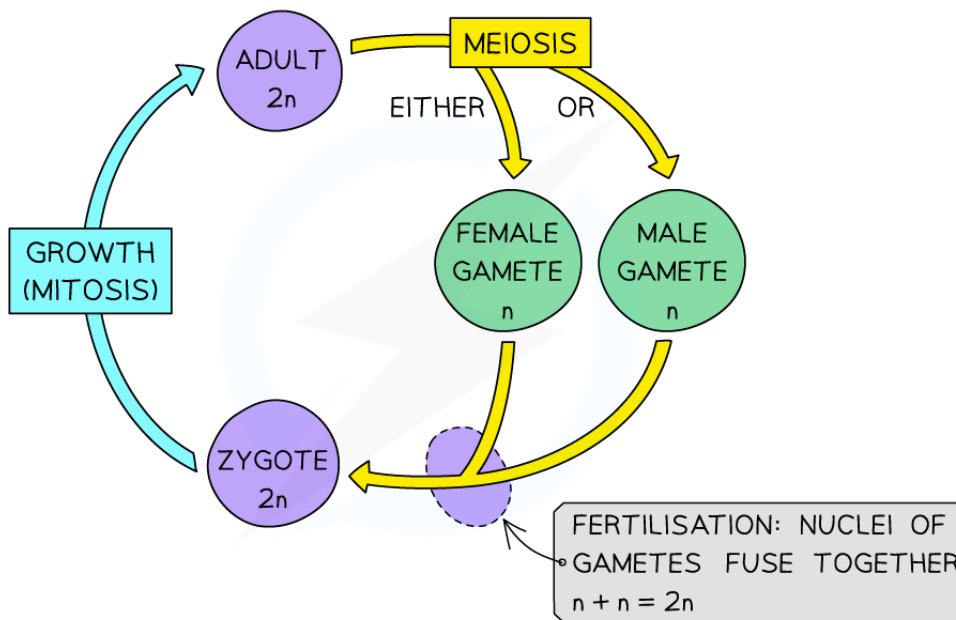
Your notes

- In the 19th century, microscopes were developed that could be used to view certain **internal cell structures**
- Around this time, it was also discovered that certain **dyes** could be used to **stain** (and **observe**) the **cell nucleus**
- The use of these dyes eventually led to the discovery of **thread-like structures** inside **dividing nuclei**
  - These were name **chromosomes**

- In the 1880s, a group of German biologists used these new developments to make detailed observations of dividing nuclei
  - Their careful observations led to **the discovery of the process of meiosis** and a basic understanding of how it occurs

- One key observation was made by viewing the chromosomes in specific cells in an organism known as the horse threadworm (*Parascaris equorum*)
  - It was observed that the **nuclei** of their **egg** and **sperm** cells contained **two chromosomes**, whereas the nuclei of a **fertilised egg** contained **four chromosomes**
  - This suggested that the chromosome number had been doubled by the process of fertilisation
  - This led to the **hypothesis** that, at some point in every generation, a **special type of nuclear division** must occur that **halves the chromosome number**

- The **specific sequence of events in meiosis** was finally discovered by carefully observing cells from the **ovaries** of **European rabbits** (*Oryctolagus cuniculus*) between 0 and 28 days old
  - This was possible because in females of this species, certain cells in the ovaries start undergoing meiosis **from birth** and the process continues **slowly** over a period of **many days**

- The initial discovery of meiosis (as well as the following series of discoveries that revealed to scientists how it occurs) was made possible through **careful scientific observations**
- Such careful observations are needed in order to **validate** the **claims** and **discoveries** that scientists make, as **scientific evidence** is required. Careful observations can enable scientists to collect **evidence that allows theories to be developed**
- The use of apparatus, dyes, sensors, amongst many other scientific tools, allows scientists to make these **precise and accurate observations**
- The methods used to make such observations must be able to be **repeated** so that the results can be confirmed by other scientists if necessary

Sexual Life Cycle

- The life cycles of organisms can be **sexual** or **asexual** (some organisms are capable of both)
  - In an **asexual** life cycle, the **offspring are genetically identical to the parent** (they have exactly the same chromosomes)
  - In a **sexual** life cycle, the **offspring are genetically distinct from each other and from each of the parents** (their chromosomes are different, causing them to be genetically distinct)

- The **halving of the chromosome number** during meiosis is very important for a **sexual life cycle** as it allows for the **fusion of gametes**
- Sexual reproduction is a process involving the **fusion** of the nuclei of two gametes to form a **zygote** (fertilised egg cell) and the production of offspring that are genetically distinct from each other
- This **fusion of gamete nuclei** is known as **fertilisation**
  - Fertilisation **doubles** the number of chromosomes each time it occurs
  - This is why it is essential that the chromosome number is also **halved** at some stage in organisms with a **sexual life cycle**, otherwise the chromosome number would keep doubling every generation
  - This halving of the chromosome number occurs during **meiosis**
  - In animals, this halving occurs during the **creation of gametes**


Sexual life cycle

3.2.2 Stages of Meiosis

DNA Replication before Meiosis

- **Before meiosis occurs**, all of the DNA inside the nucleus of the 'parent' cell is **replicated**
  - This occurs during a period of the cell cycle known as **interphase**

- Once this has occurred, each chromosome now consists of **two genetically identical sister chromatids**, which are joined together by a **centromere**
  - The sister chromatids are genetically identical because DNA replication is a very accurate process and only a very small number of mistakes occur when DNA is being copied

- The two DNA molecules formed by DNA replication prior to meiosis are considered to be **sister chromatids** until the **splitting of the centromere** at the start of **anaphase** (a stage during meiosis II, during which the sister chromatids are **pulled apart**)
- After this, they are once again considered as **individual chromosomes**

Your notes
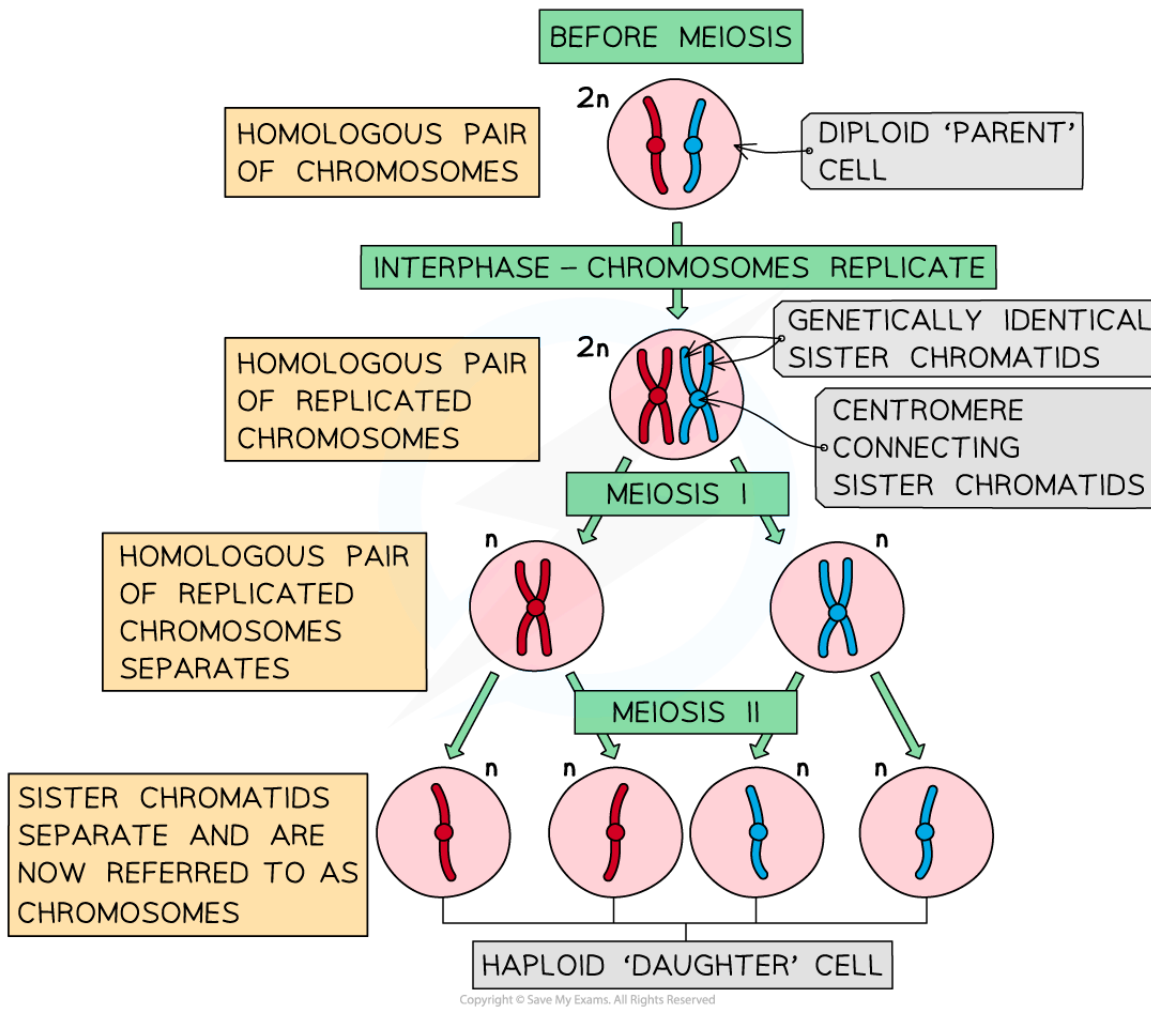
DNA replication before meiosis


💡 **Examiner Tip**

Understanding the difference between chromosomes and chromatids can be difficult. We count chromosomes by the **number of centromeres present**. So when the 46 chromosomes duplicate during interphase and the **amount of DNA in the cell doubles** there are still only 46 chromosomes present because there are still only 46 centromeres present. However, there are now 92 chromatids, which are strands of replicated chromosomes.

Formation of Bivalents & Crossing Over

- At the start of meiosis, **homologous chromosomes pair up** with each other
  - As DNA replication has already occurred, each chromosome is made up of **two sister chromatids**
  - This means that a pair of homologous chromosomes is made up of **four DNA molecules**

- A pair of homologous chromosomes is known as a **bivalent**
- The pairing process resulting in the formation of a bivalent is known as **synapsis**
- After synapsis has occurred, a process known as **crossing over** may occur
- During crossing over, two **non-sister chromatids** (i.e. one chromatid from each of the homologous chromosomes) form a **junction**
- At this junction, the two chromatids **break** and **rejoin** with each other
- As these crossover events occur at exactly the same position on the two non-sister chromatids, this allows **genes** to **exchange** between the chromatids
- Non-sister chromatids are **homologous** but are **not genetically identical** and this means that some of the **alleles** of the exchanged genes will be **different**
- This process, therefore, produces chromatids with **completely new combinations of alleles** (that were not previously present in the DNA of the 'parent' cell)
- As these chromatids will eventually be split up into **different gametes**, crossing over is of great **importance** because it is a significant source of **genetic variation** between gametes
  - This ensures there is genetic variation in **populations** of **sexually-reproducing species**, which is key to a species' ability to **evolve** and **adapt** to changes in its environment over time
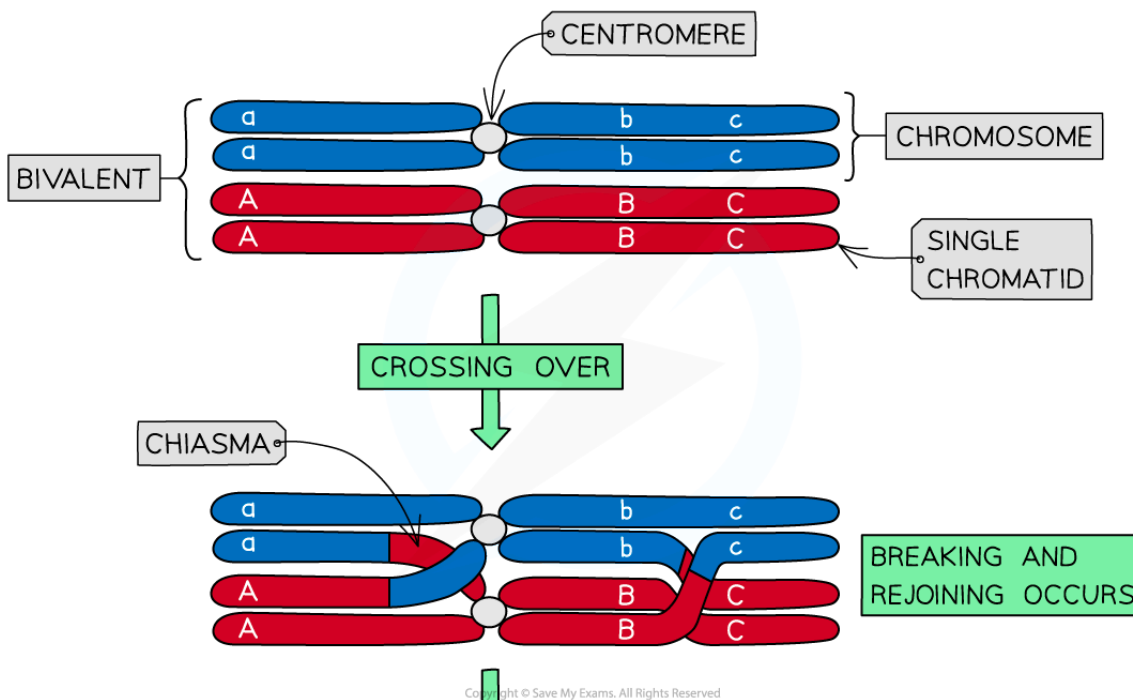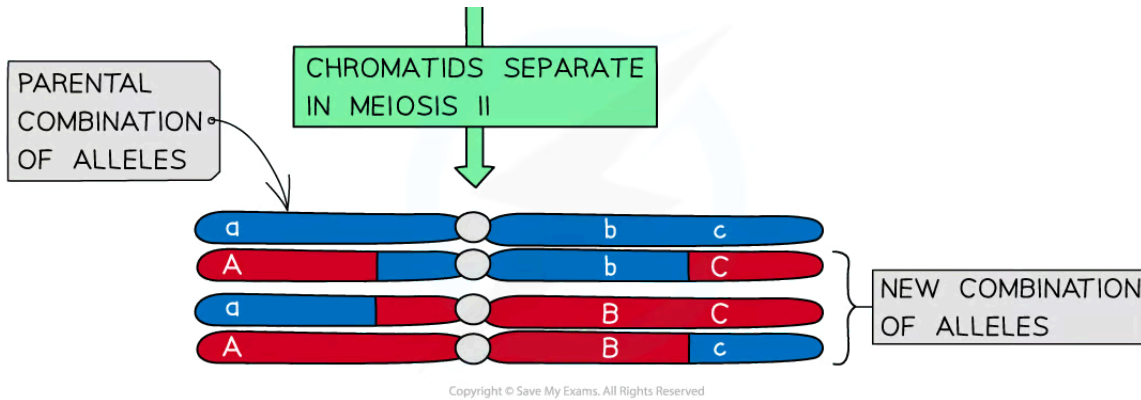
Crossing over of non-sister chromatids leading to the exchange of genetic material

Random Orientation

- At metaphase, during meiosis I, **homologous chromosomes line up at the cell equator** as they prepare to **separate**
- **Spindle microtubules** grow out from the **poles** of the cell and attach to the **centromeres** of the chromosomes
- Each of the two homologous chromosomes in a bivalent is attached to a **different pole**
- The **orientation of the bivalents** when they line up at the cell equator determines which pole each chromosome gets attached to (and eventually pulled towards)
- The orientation of the bivalents is **completely random**
- In addition, the bivalents also **assort independently of one another** (i.e. the orientation of one bivalent never affects the orientation of another)
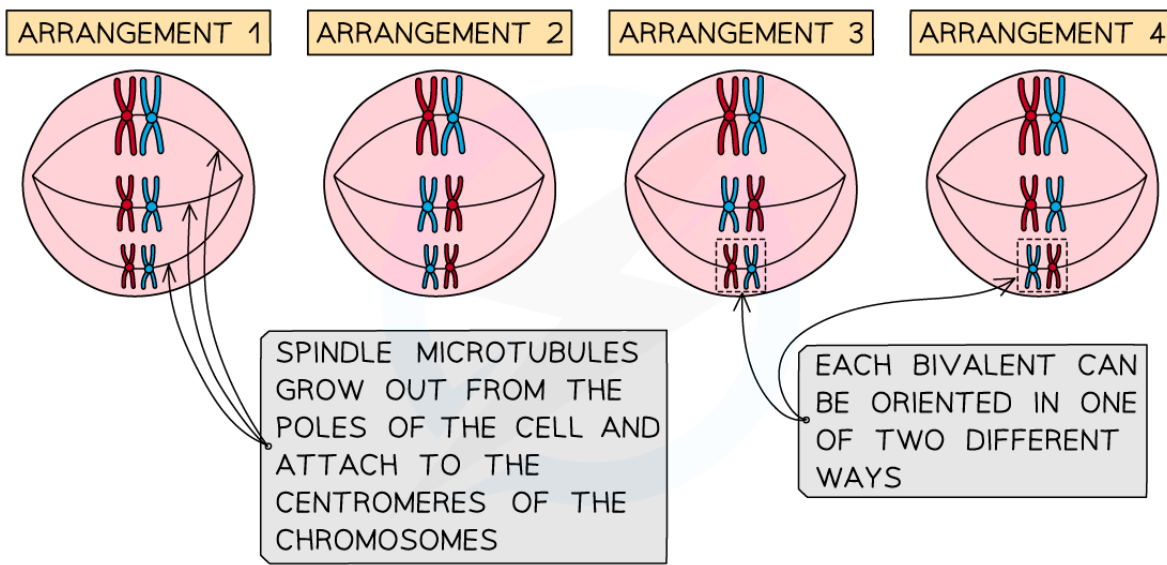
The orientation of bivalents lining up at the cell equator is random

Reduction Division

- During meiosis, the homologous chromosomes forming a bivalent separate in a process known as **disjunction**
- The homologous chromosomes then move to **opposite poles** of the cell
- As **one chromosome of each type** moves to each pole, the two separate nuclei formed by the first division of meiosis (meiosis I) now only contain **one of each type of chromosome**, making the two new cells **haploid**
  - Essentially, the chromosome number of the cells has been **halved**

- This is why the first division of meiosis is known as a **reduction division**
  - The chromosome number has been **reduced** (halved) from **diploid** to **haploid**

3.2.3 Genetic Variation

Your notes

Genetic Variation & Meiosis

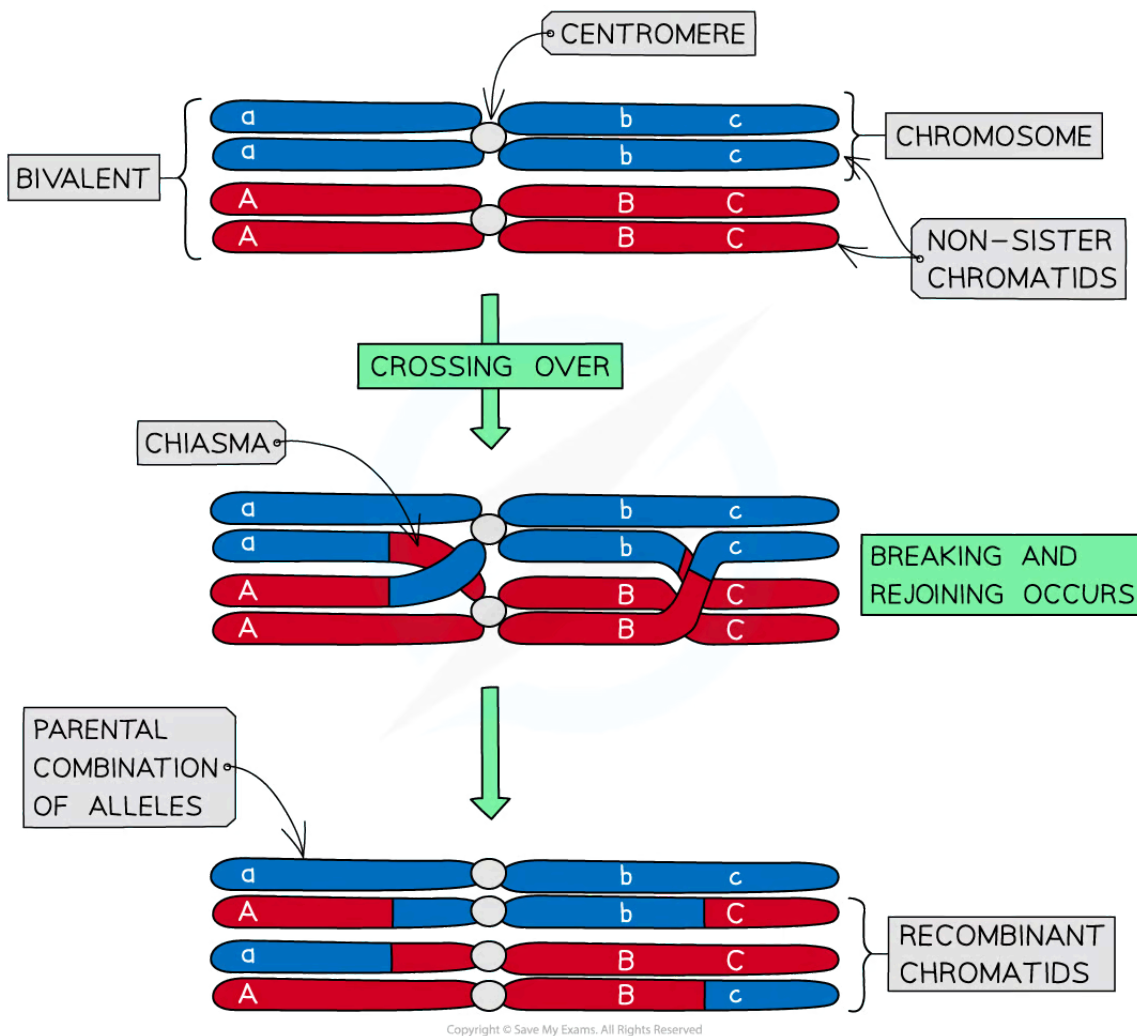### Crossing over and random orientation promote genetic variation

- Having **genetically different offspring** can be **advantageous** for natural selection and therefore increase the survival chances of a species
- **Meiosis** has several mechanisms that **increase the genetic variation of gametes** produced
- Both **crossing over** and **random orientation** result in **different combinations of alleles** in gametes

Crossing over

- Crossing over is the process by which **non-sister chromatids exchange alleles**
- Process:
  - During prophase I of meiosis homologous chromosomes pair up and are in very close proximity to each other
    - A pair of homologous chromosomes can be referred to as a **bivalent**

  - At this point, there can be an exchange of genetic material **(alleles)** between **non-sister chromatids** in the bivalent
  - The crossing points are called **chiasmata**
  - This results in a **new combination of alleles on the two chromosomes** (these can be referred to as **recombinant chromosomes**)

- This swapping of alleles is a significant source of genetic variation because it can occur at multiple random positions along the chromosome
- Crossing over can happen anywhere along the chromosome but is more likely to occur further down the chromosome away from the centromere

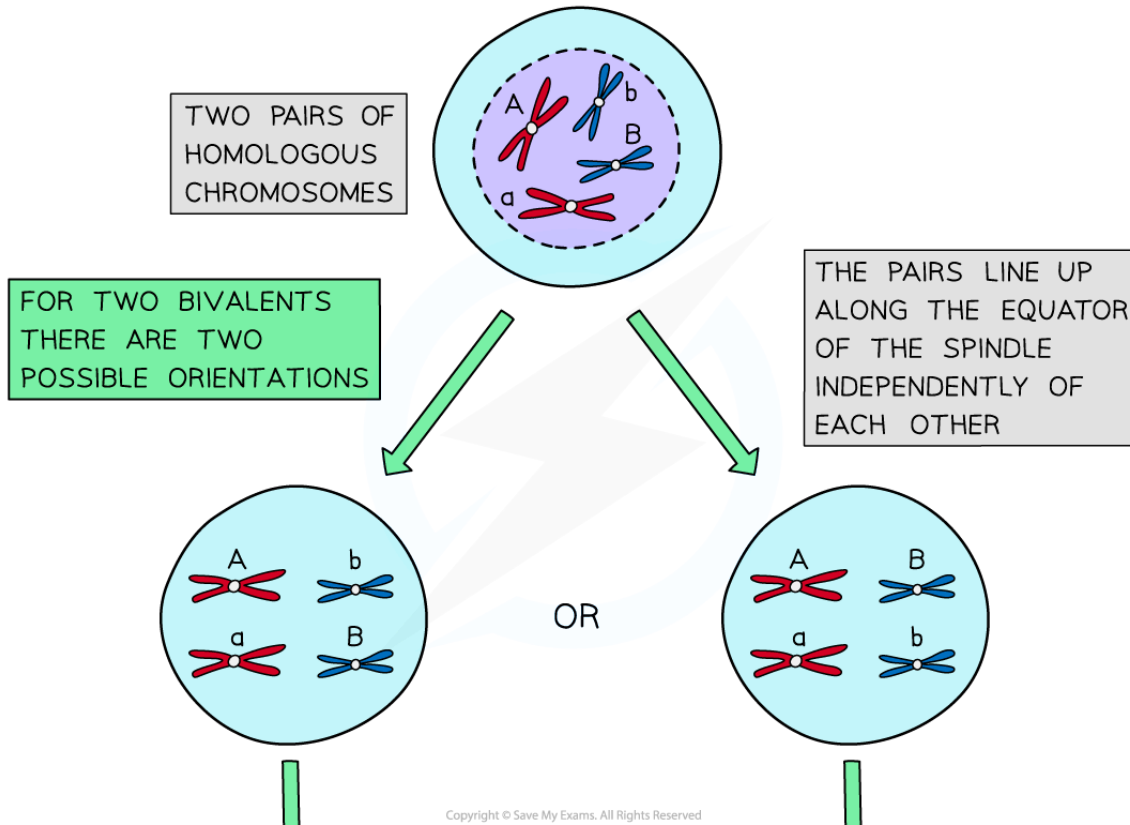*Crossing over occurring between two non-sister chromatids*

Random orientation

- The **random orientation of homologous pairs along the equator of the cell** during **metaphase I** result in the production of **different allele combinations in daughter cells**
- In prophase I, homologous chromosomes pair up and in metaphase I, they align along the equator of the cell
  - Each pair can be arranged with **either chromosome on top**, this is completely random
  - The **orientation of each homologous pair** is **random/independent** (unaffected by the orientation of any other pair)

- In anaphase I the homologous chromosomes are **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

- The different combinations of chromosomes in daughter cells increases genetic variation between gametes
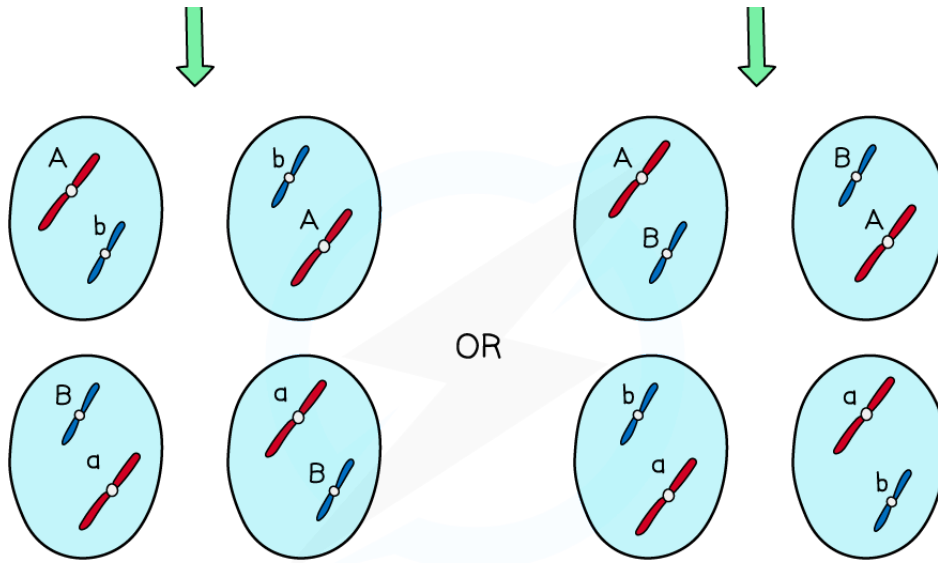
AT THE END OF MEIOSIS II, EACH ORIENTATION GIVES TWO TYPES OF GAMETE. THERE ARE THEREFORE FOUR TYPES OF GAMETE ALTOGETHER

The random orientation of homologous chromosomes leads to different genetic combinations in daughter cells

The different combinations of chromosomes following meiosis

- The number of possible chromosomal combinations resulting from random assortment is equal to 2^n
  - n is the number of homologous chromosome pairs or haploid number

- For humans: the number of chromosomes is 46 meaning the number of homologous chromosome pairs is 23 so the calculation would be:
  - 2^{23} = 8,388,608 possible chromosomal combinations

✏️ **Worked example**

Calculate how many different chromosomal combinations can result from meiosis in a plant species which has a diploid number of 16. Assume no crossing over occurs.

[1 mark]

Step 1: Use the relevant formula

$$2^n$$

Step 2: Calculate the haploid number

Diploid number (2n) = 16

Haploid number (n) = $16 \div 2 = 8$

Step 3: Substitute in figures

$2^8 = 256$

There are **256** different chromosomal combinations that can occur.

Genetic Variation & Fertilisation

Fusion of gametes from different parents promotes genetic variation

- **Meiosis** creates genetic variation **between the gametes** produced by an individual through crossing over and random orientation
- This means each gamete carries substantially **different alleles**
- During fertilisation, any male gamete can fuse with any female gamete to form a zygote
- This **random fusion of gametes** at fertilisation creates genetic variation **between zygotes** as each will have a unique combination of alleles

The different combinations of chromosomes following fertilisation

- In random fertilisation, **any two gametes may fuse together**
- Therefore the formula to calculate the number of combinations of chromosomes after the random fertilisation of two gametes is **$(2^n)^2$**
  - n is the haploid number and ^{2} is the number of gametes
  - Therefore in humans, when the haploid number is 23, the number of combinations following fertilization is $(2^{23})^2$ = 70,368,744,177,664

- This explains why relatives can differ so much from each other. Even with the same parents, individuals can be genetically distinct due to variation at the meiosis and fertilization stage (as well as other possible mutations and crossing-over)

✏️ Worked example

Calculate the number of different possible chromosome combinations after the random fertilization of an ovule and pollen nuclei from the same plant species (Diploid number = 16).

[2 marks]

Step 1: State formula for random fertilisation between any two gametes

$$(2^n)^2$$

Step 2: Use information from question to state haploid number

$$n = 8$$

Step 3: Substitute in figures

$$(2^n)^2$$

$$(2^8)^2$$

Answer **65,536**

💡 Examiner Tip

These sources of genetic variation explain why relatives can differ so much from each other. Even with the same parents, individuals can be genetically distinct due to the processes outlined above.While we can calculate the number of chromosomal combinations that result from random orientation and random fertilisation, the number of combinations from crossing over is infinite, or as good as!

Your notes

3.2.4 Non-disjunction

Non-disjunction

- **Non-disjunction** occurs when chromosomes fail to separate correctly during **meiosis**
- This can occur in either anaphase I or anaphase II, leading to **gametes** forming with an **abnormal number of chromosomes**
  - The gametes may end up with **one extra copy** of a particular chromosome or **no copies** of a particular chromosome
  - These gametes will have a different number of chromosomes compared to the normal haploid number

- If the abnormal gametes are fertilized, then a **chromosome abnormality** occurs as the diploid cell (**zygote**) will have the incorrect number of chromosomes
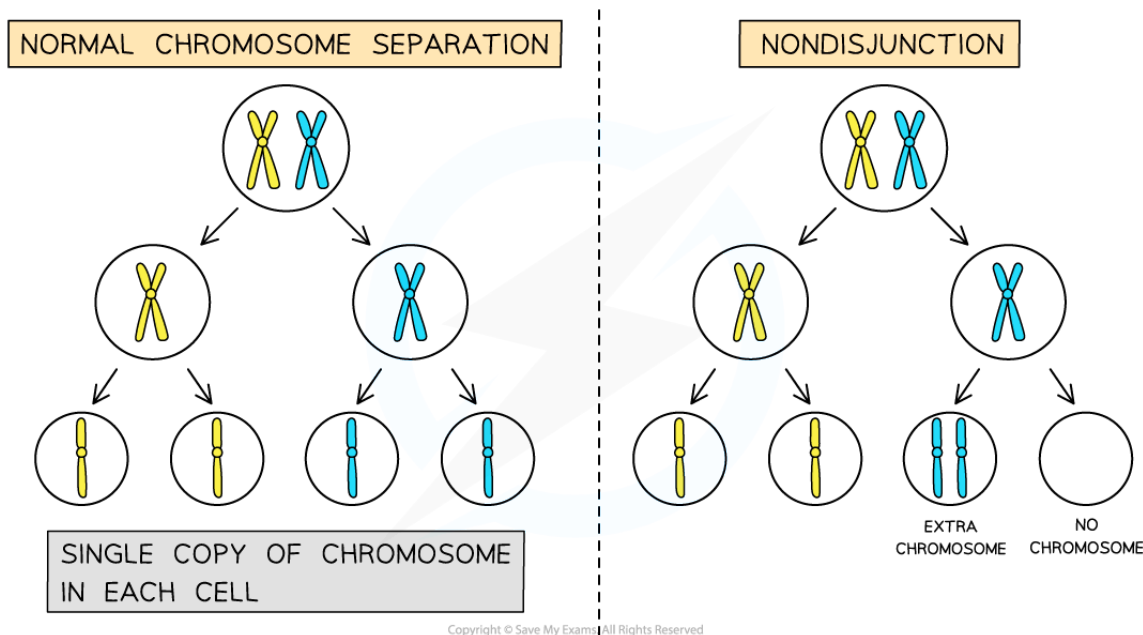
Image showing how chromosomes failing to separate properly during meiosis can result in gametes with the incorrect number of chromosomes

Down Syndrome

- A key example of a non-disjunction chromosome abnormality is Down syndrome, also called **Trisomy 21**
- Non-disjunction occurs during anaphase I (in this case) and the 21st pair of homologous chromosomes fail to separate
- Individuals with this syndrome have a total of 47 chromosomes in their cells as they have three copies of **chromosome 21**

Your notes

- The impact of trisomy 21 can vary between individuals, but some common features of the syndrome are **physical growth delays** and **reduced intellectual ability.** Individuals can also suffer from issues with **sight** or **hearing**

Other trisomy syndromes

- There are other trisomy possibilities that can result from non-disjunction; many, but not all, have very serious impacts on the phenotype of the offspring which may be fatal
  - **Patau syndrome** (trisomy 13) and **Edwards syndrome** (trisomy 18) are very serious syndromes which result in many physical disabilities and developmental difficulties
    - Trisomy 18 and 13 both have **very low survival rates** with few babies surviving past their first birthday

  - **Klinefelter's syndrome** is caused by non-disjunction in sex chromosomes which leads to having the chromosomes XXY
    - This syndrome is often not diagnosed until adulthood and doesn't impact life expectancy but may have a negative effect on fertility

  - **Turners syndrome** also affects the sex chromosomes with individuals possessing just one X chromosome
    - Individuals with Turners syndrome would not necessarily have a reduced life-expectancy, although will often be shorter and may suffer some symptoms such as lack of sexual development during puberty

Age & Non-disjunction

- Many studies have shown that there is a correlation between age and the incidence of non-disjunction
- It is believed that as the **age of the parents increases** the **incidence of non-disjunction increases**
- In particular, the age of the **mother** has been found to increase the chance of having a child with Down Syndrome
  - The impact of age on the risks is represented in the table below

Age and Risk of Down's Syndrome Table

| Mother's age (Years)	25−29	30−34	35−39	40−44	45+
Chance of having baby with Down Syndrome	1 in 1250 live births	1 in 1000 live births	1 in 400 live births	1 in 100 live births	1 in 30 live births

Methods used for Karyotype Analysis

Methods can be used to obtain cells for karyotype analysis

- A **karyotype** can be created to show an image of all of the chromosomes of an individual from a single cell.
- Chromosomes are arranged into their homologous pairs and studied to check for any abnormalities
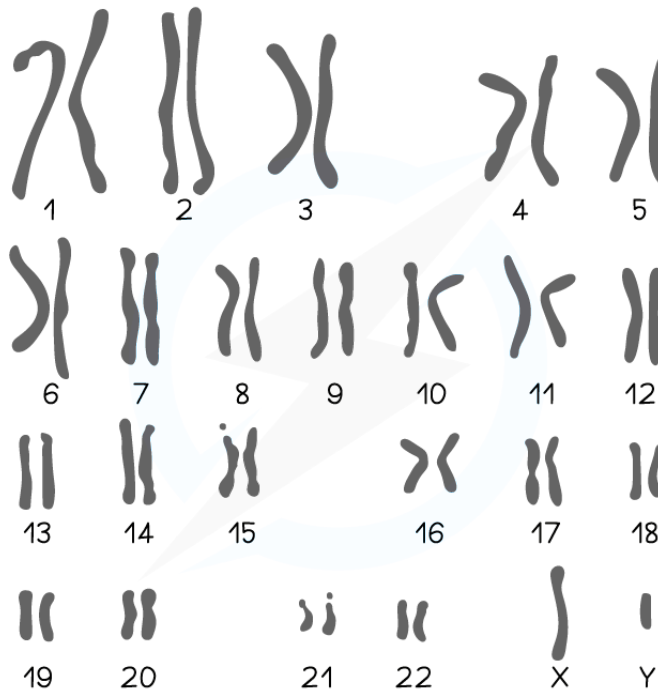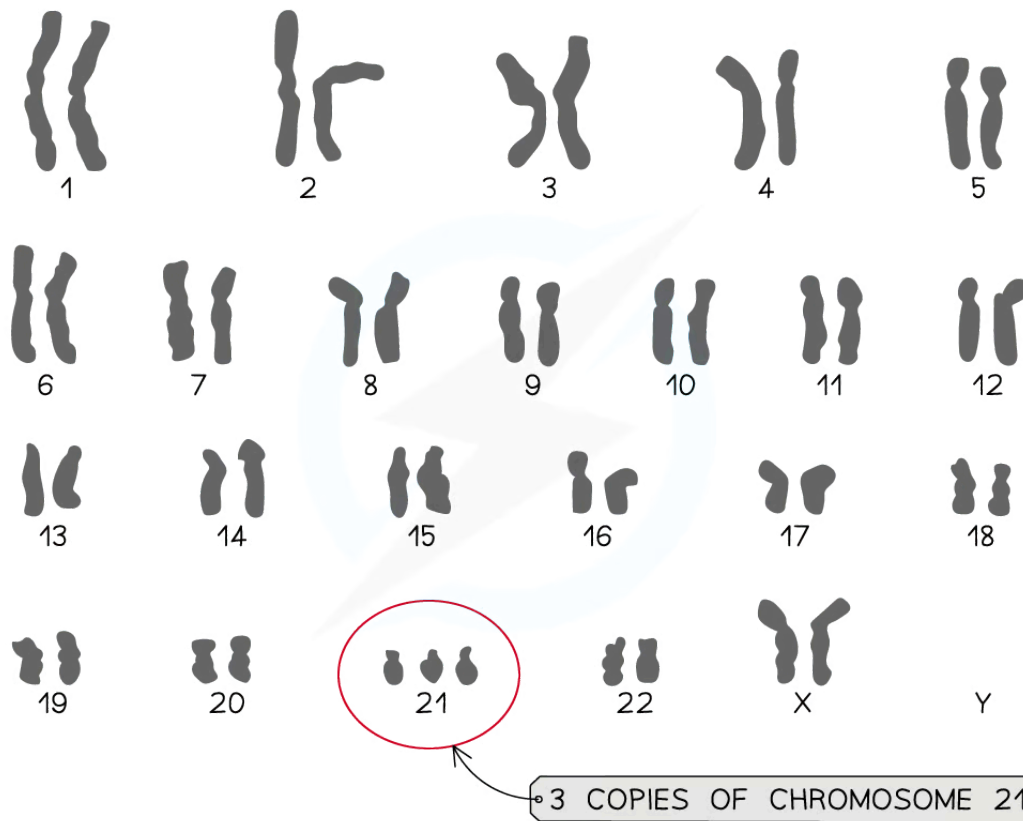

This karyogram shows a typical or "normal" karyotype

- A karyotype which shows a chromosomal abnormality may have incorrect numbers of chromosomes present
- Trisomy syndromes will show a third chromosome present at one of the chromosome positions
  - For example, Down's syndrome shows a third chromosomes at the 21st position

Your notes

Your notes

A karyogram showing the karyotype of an individual with Down's Syndrome

- Two methods can be used to obtain cells from an **unborn child** for chromosome testing:
  - **Amniocentesis**
  - **Chorionic villus sampling**

Amniocentesis

- A needle is inserted through the mother's abdomen wall and a small sample of **amniotic fluid** is taken. The sample will contain some foetal cells for analysis
- This procedure usually takes place around 16 weeks of pregnancy
- There is a **small risk of miscarriage** associated with the procedure (approx 1%)
- The procedure also poses a small risk of infection

Chorionic Villus Sampling (CVS)

- A long tube is inserted through the vagina and then cervix in order to take a small sample of the developing **chorion** (a membrane surrounding the embryo which forms part of the placenta)
- This procedure can be carried out earlier in the pregnancy; around 10 - 12 weeks
- CVS has a slightly **increased risk of miscarriage** associated with the procedure (approx 2%)
- There is also a small risk of infection

3.2.5 Skills: Meiosis

## Drawing the Stages of Meiosis

- Cells undergoing meiosis can be observed and photographed using specialized microscopes
- The different stages of meiosis have distinctive characteristics meaning they can be identified from photomicrographs
- Being able to identify the stages of meiosis from photomicrographs and diagrams is an important skill for a biologist

Step 1: Identifying if meiosis I or meiosis II is occurring

- **Homologous chromosomes** pair up side by side in meiosis I only
  - This means if there are **pairs of chromosomes** in a diagram or photomicrograph **meiosis I** must be occurring

- The **number of cells forming** can also help identify whether meiosis I or II is occurring
  - If there are **two new cells** forming it is **meiosis I** but if there are **four new cells** forming it is **meiosis II**

Identifying which stage of meiosis I is occurring

- **Prophase I**: **Homologous pairs** of chromosomes are visible in diploid cell (2n). Crossing over occurs
- **Metaphase I**: Spindle fibres pull homologous pairs so they are lined up **side by side** along the **equator** of the cell. Orientation of homologous chromosomes is random
- **Anaphase I**: **Whole chromosomes** are being pulled to opposite **poles** with **centromeres intact**
- **Telophase I:** There are **2 groups** of condensed chromosomes around which nuclei membranes are forming
- **Cytokinesis:** Cytoplasm is dividing and the **cell membrane is pinching inwards** to form **two cells** with haploid chromosome numbers (n)
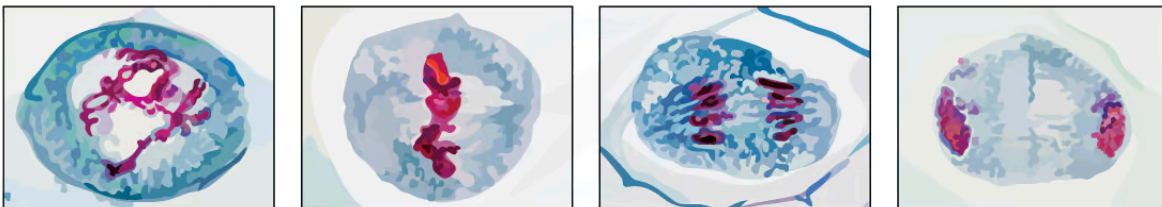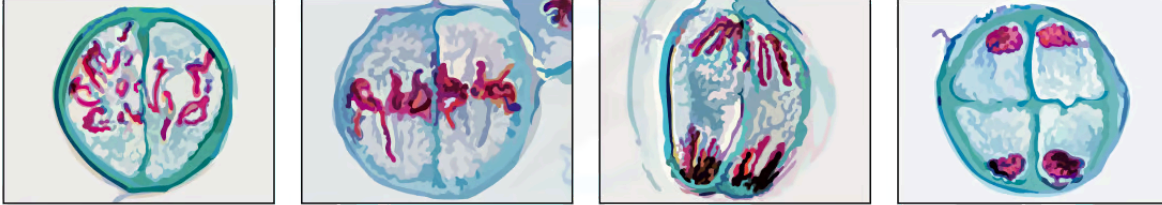
Prophase I, Metaphase I , Anaphase I and Telophase I as seen in photomicrographs

Identifying which stage of meiosis II is occurring

- **Prophase II**: **Single whole chromosomes** are visible in haploid cells
- **Metaphase II**: Single whole chromosomes are lined up along the **equator** of the cell in a **single file**
- **Anaphase II**: **Centromeres divide** and **chromatids** are being pulled to opposite **poles**
- **Telophase II:** Nuclei are forming around the **4 groups** of condensed chromosomes

Cytokinesis: Cytoplasm is dividing and **four haploid cells** are forming

Prophase II, Metaphase II , Anaphase II and Telophase II as seen in photomicrographs

Drawing the stages of meiosis

- The distinguishing features mentioned above can also be used by biologists to draw scientific diagrams of meiosis I and meiosis II
- The conventions for drawing are:
  - The drawing must have a title
  - A **sharp HB pencil** should be used (and a good eraser!)
  - Drawings should be on plain white paper
  - Lines should be **clear**, **single lines** (no thick shading)
  - **No shading**
  - The drawing should take up as much of the space on the page as possible
  - Well-defined structures should be drawn
  - The drawing should be made with **proper proportions**
  - **Label lines** should not cross or have arrowheads and should **connect directly** to the part of the drawing being labelled
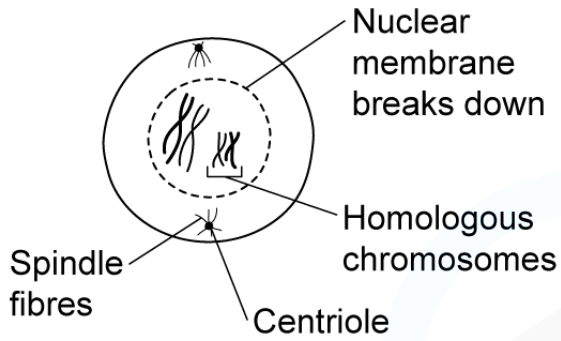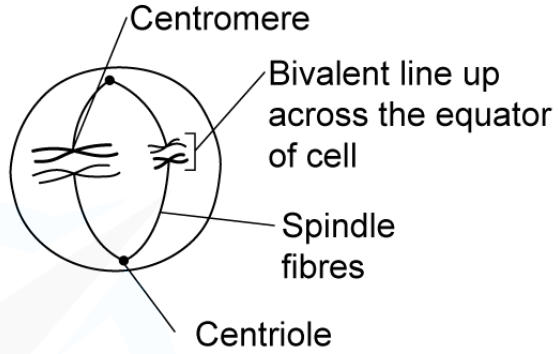
Prophase I

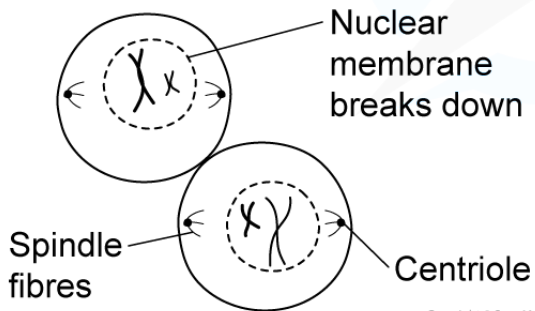Nuclear membrane breaks down

Homologous chromosomes

Spindle fibres

Centriole

Metaphase I

Centromere

Bivalent line up across the equator of cell

Spindle fibres

Centriole

Prophase II

Nuclear membrane breaks down

Spindle fibres

Centriole

Metaphase II

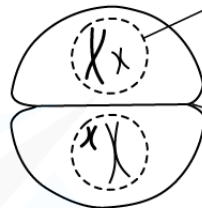
Chromosomes line up along equator of cell

Your notes

Anaphase I

Spindle fibres contract and pull chromosomes to poles of cell

Telophase I
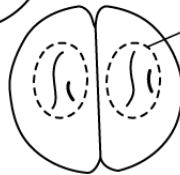
Nuclear membrane forms

Cytokinesis begins

Anaphase II

Sister chromatids pulled to poles of cell

Telophase II

4 haploid daughter cells

Nuclear membrane forms

Cytokinesis

Drawing the different stages or phases of meiosis

> 💡 **Examiner Tip**
>
> For metaphase remember **M for the middle** of the cell which is where the chromosomes will be lined up. For anaphase remember **A for away** from the middle to the poles, which is where the chromosomes / chromatids are being pulled. When drawing the stages of meiosis you do not have to show crossing over occurring.