IB Biology DP

YOUR NOTES ↓

10. Genetics & Evolution (HL Only)

CONTENTS

YOUR NOTES

10.1 Meiosis

10.1.1 The Process of Meiosis

Replication of Chromosomes

Like mitosis, meiosis is preceded by DNA replication

- Like most other eukaryotic cells, gamete-producing cells perform a **cell cycle**
  - Gamete-producing cells are found in the testes and ovaries
    - The cells that give rise to sperm cells are found in the testes
- If a new sperm-producing cell is being generated in the testes, **mitosis** occurs as with any other diploid body cell
  - The G1, S and G2 stages of the cell cycle proceed
  - The sperm-producing cell undergoes **DNA replication** and the amount of DNA within that cell doubles
  - The cell still contains **2n** number of chromosomes; although each chromosome has doubled in size
- DNA replication also happens before **meiosis**
  - Unlike mitosis, replication in meiosis is followed by **2 rounds** of chromosome separation as opposed to **1 round in mitosis**
- **Hormones** and **other stimuli** trigger cells in the testes to enter **meiosis;** at this stage the sperm-producing cell **ceases to be diploid** and fulfils its function to produce **haploid gametes** (spermatids, which then develop into spermatozoa, also known as sperm cells)
  - The triggering of **ova generation** in female mammals is **less well understood** because of the different times of a female mammal's life when eggs are produced
    - Male mammals tend to produce sperm throughout their adult life

Mitosis or Meiosis?

- Gamete-producing cells are unique in that they can divide by **both well-known cell division routes**
- Considering a sperm-producing cell as an example, it has two possible routes of cell division
  - Mitosis - to **regenerate** itself and during **growth** of the tissue in the testes
    - Like any other somatic cell in this regard
  - Meiosis - to fulfil its specialisation ie. to produce **sperm** (called spermatogenesis) at the required time of the male's life
- Both routes begin with **DNA replication** within the diploid cell's nucleus

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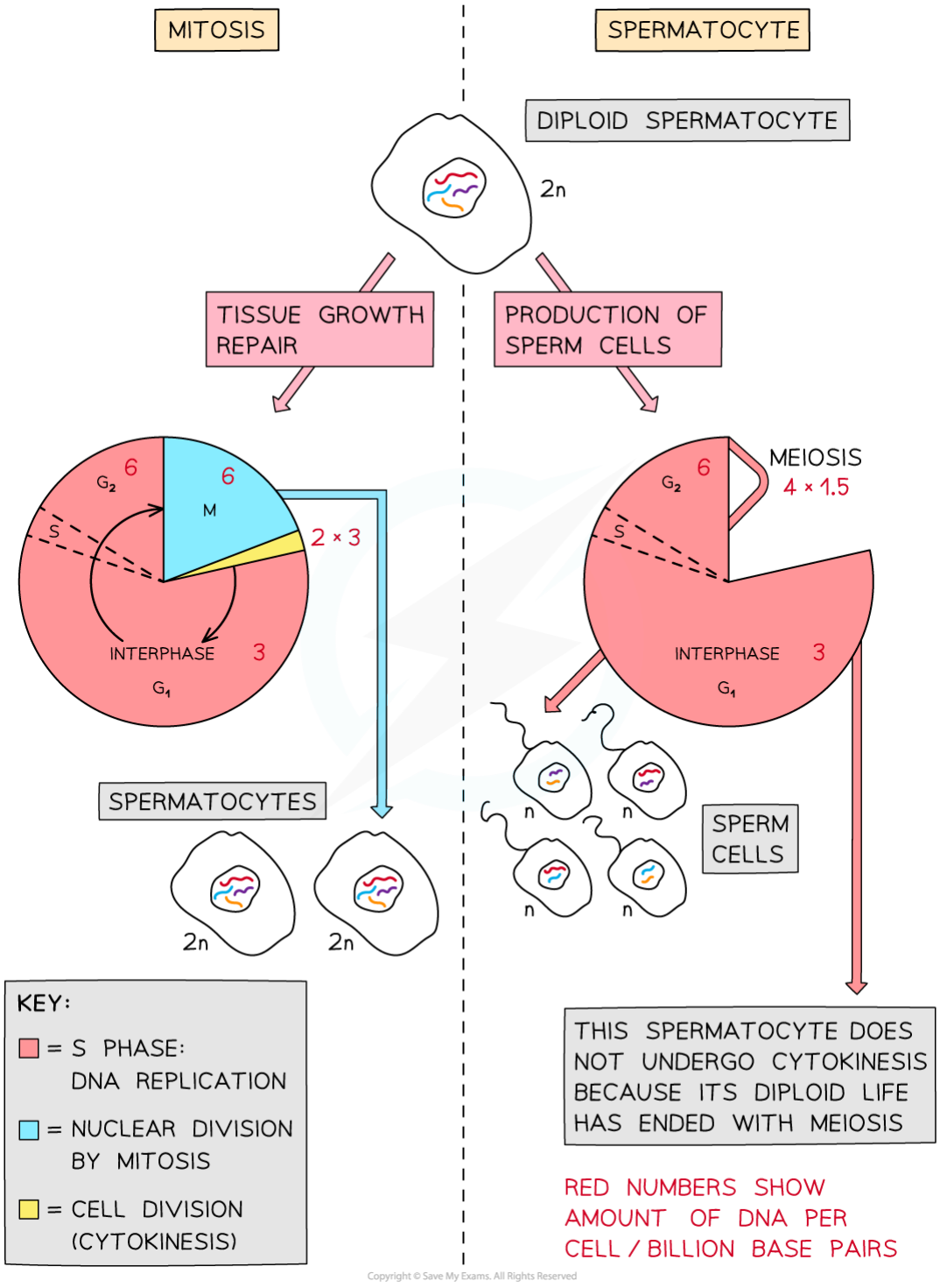

The two possible routes of cell division for a spermatocyte. Both start with DNA replication. The right hand side describes spermatogenesis

DNA Replication

- During interphase, the cell increases in mass and size and carries out its **normal cellular functions**

YOUR NOTES

- eg. **synthesising proteins** and the reactions of **respiration**
- Interphase consists of three phases:
  - G1 phase
  - S phase
  - G2 phase
- During interphase the DNA in the nucleus **replicates,** after which each chromosome consists of two identical sister **chromatids**
  - This phase of interphase during which DNA replication occurs is called the S phase – S stands for **s**ynthesis (of DNA)

Following DNA replication, the fate of the sperm-producing cell is determined by hormonal and other stimuli

- If the male is sexually mature, **some cells in the testes will enter meiosis** and begin producing sperm
  - The individual **sperm-producing cell ceases to exist** when it enters meiosis
- Other sperm-producing cells will divide by mitosis
  - To ensure a **healthy population** of sperm-producing cells for future sperm production
  - A sperm-producing cell retains its identity by this route

The Process of Meiosis

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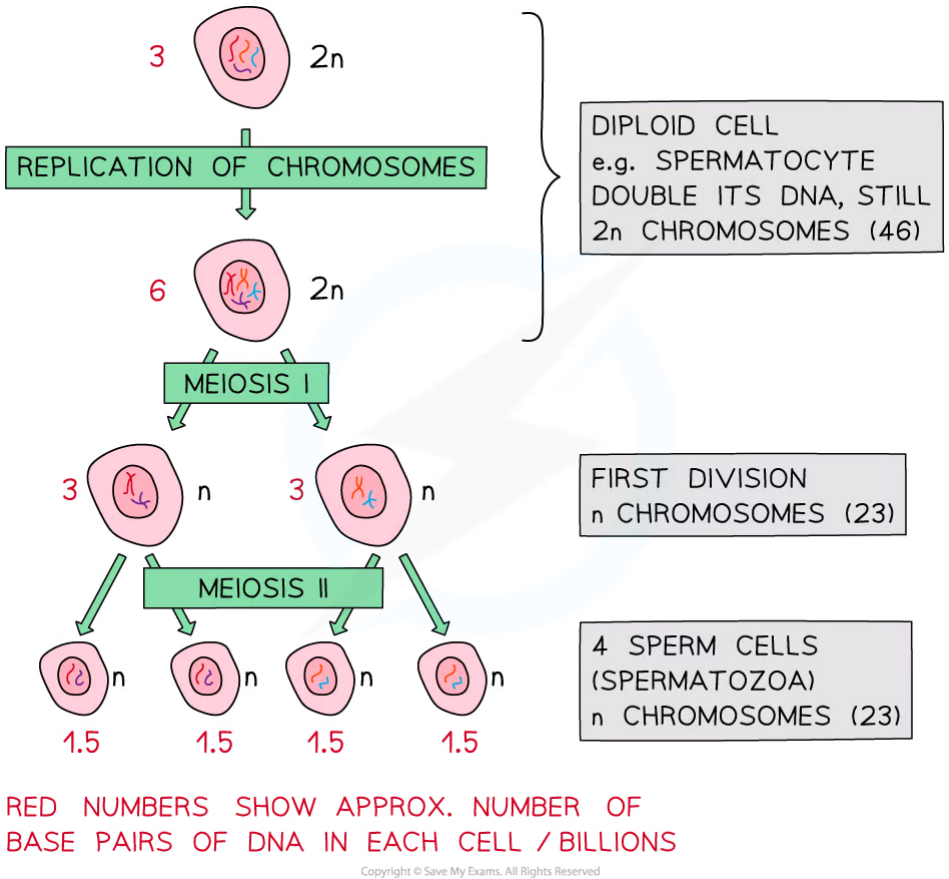
Overview of Meiosis

- Meiosis is a form of nuclear division that results in the production of haploid cells from diploid cells
- It produces gametes in plants and animals that are used in **sexual reproduction**
- It has many similarities to mitosis but it has two successive divisions: **meiosis I** and **meiosis II**
- As with mitosis, within each division there are four stages; prophase, metaphase, anaphase and telophase
- Meiosis occurs:
  - In the **testes** of male animals and the **ovaries** of female animals
  - In the **anthers** and **ovaries** of flowering plants
- Meiosis leads to the production of the following haploid gametes:
  - **Spermatozoa,** or **sperm cells,** in male animals, **ova** (singular ovum) in female animals
  - Male plant gametes are carried in **pollen** grains and female plants gametes are held in the **ovules** within the plant ovary.



***Overview of meiosis, showing chromosome numbers and quantities of DNA present in each cell in humans* (Homo sapiens)**

Formation of Bivalents

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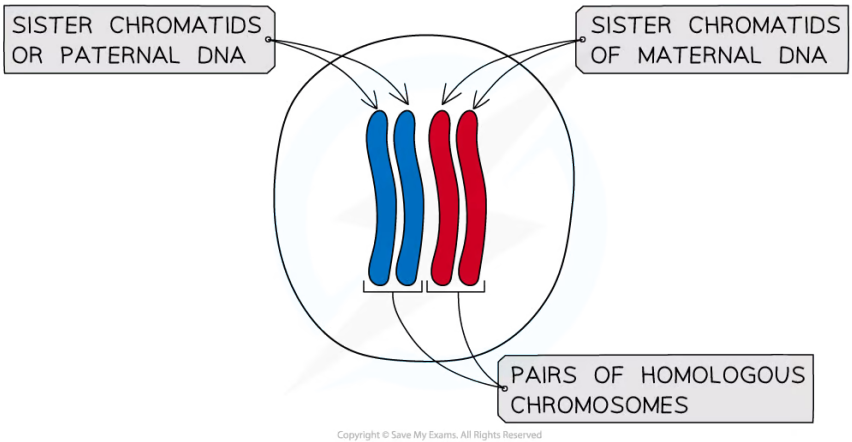
- After DNA replication, the first step of the meiotic pathway (prophase I), is that the chromosomes **condense**
  - This means that they shorten, become denser and so **become visible**
  - Condensation separates the jumble of chromatin in the nucleus and allows the chromosomes to segregate properly later, in meiosis
- Each chromosome is visible as a pair of chromatids
  - **Sister chromatids** are so-called because they originate from the same parent
    - This is **not a reference to gender/sex** chromosomes
  - Two homologous chromosomes (exact copies of each other) align alongside to each other
    - This is called a **bi**valent because it is composed of **two** chromosomes
    - It is also called a **tetra**d because it is composed of **four** chromatids
    - This process is called synapsis
  - An example of a bivalent would be for human chromosome number 11 (see image below)
  - The original chromosome pair 11 has one chromosome no. 11 inherited from the **paternal** line ie. from the organism's father and one no. 11 chromosome from the **maternal** line
    - Each chromosome 11 copies itself in interphase
      - So there are 2 identical copies of paternal chromosome 11
      - And 2 identical copies of maternal chromosome 11
    - Such a bivalent is also a **tetrad** because each of the two copies of chromosome 11 is made up of 2 chromatids, making 4 chromatids in total



Exam Tip

The cells of the sex organs can divide by both mitosis and meiosis, while other body cells (aka somatic cells) only divide by mitosis.

'Cell division' is sometimes a confusing term because that implies that DNA is being 'divided' between cells. This IS strictly true, although in order for there to be enough DNA for the new cells, DNA has to replicate itself first.

To aid fluency in exam answers, write a glossary or flashcards of the following terms so you can always choose the right the keywords in your written answers: allele; bivalent; centromere; chromosome; chromatid; gene loci; homologous pair; synapsis; tetrad.

YOUR NOTES

YOUR NOTES

Crossing Over

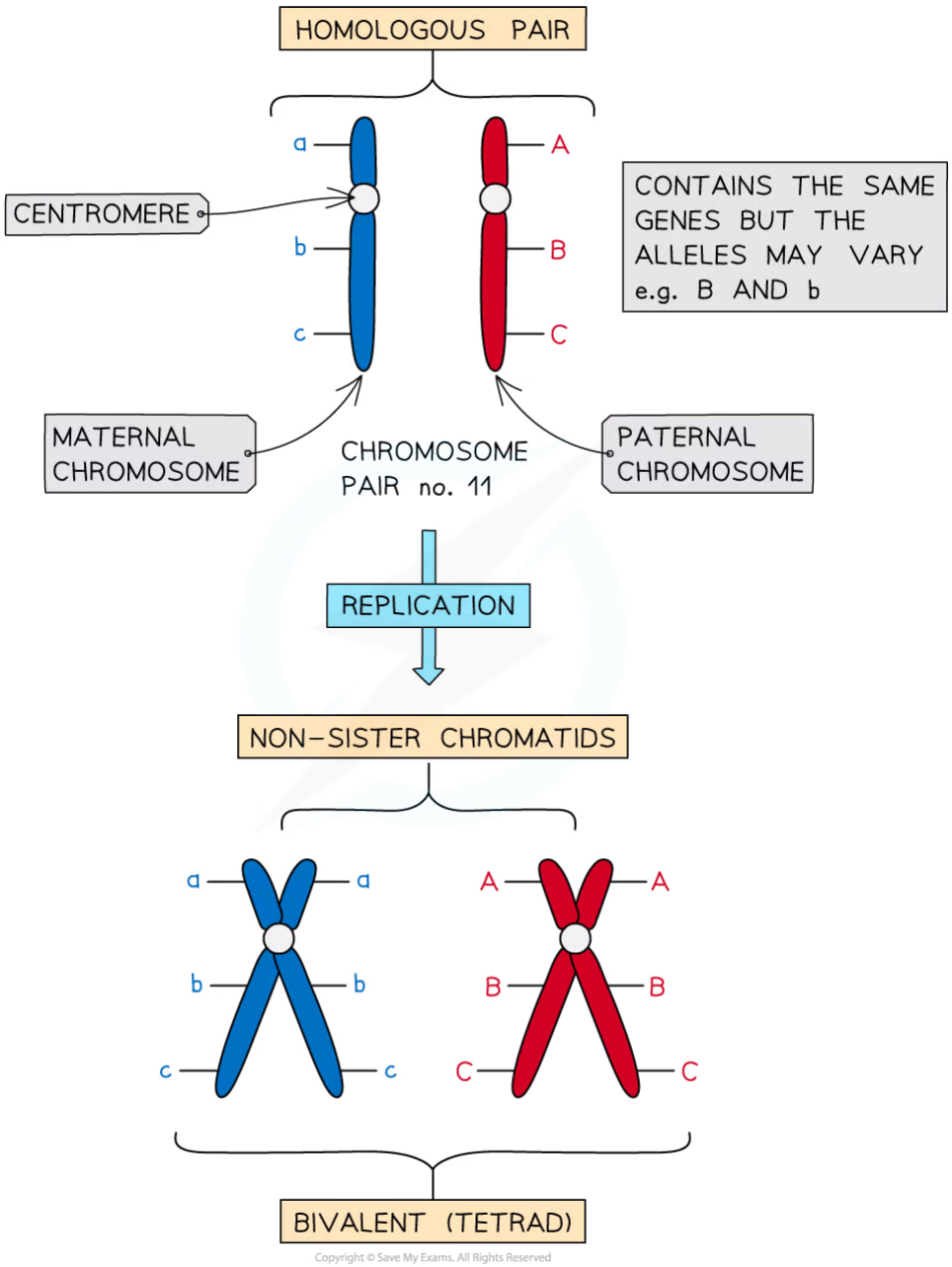
Crossing over is the exchange of DNA material between non-sister homologous chromatids

- Meiosis has several mechanisms that **increase the genetic diversity of gametes** produced
- Both **crossing over** and **independent assortment** (sometimes also called **random orientation**) result in different combinations of alleles in gametes

What are non-sister chromatids?

- In a diploid cell, each homologous pair of chromosomes consists of one chromosome that originated from the organism's father, and one from the mother
- During replication prior to meiosis, each chromosome copies to form a **bivalent**
- The **chromatids align in prophase I**, during which paternal chromatids and maternal ones can line up directly against each other
- If a pair of adjacent chromatids are originated from two different parental chromosomes, they are called **non-sister chromatids**
    - As such, they carry the same genes but can carry different alleles

YOUR NOTES



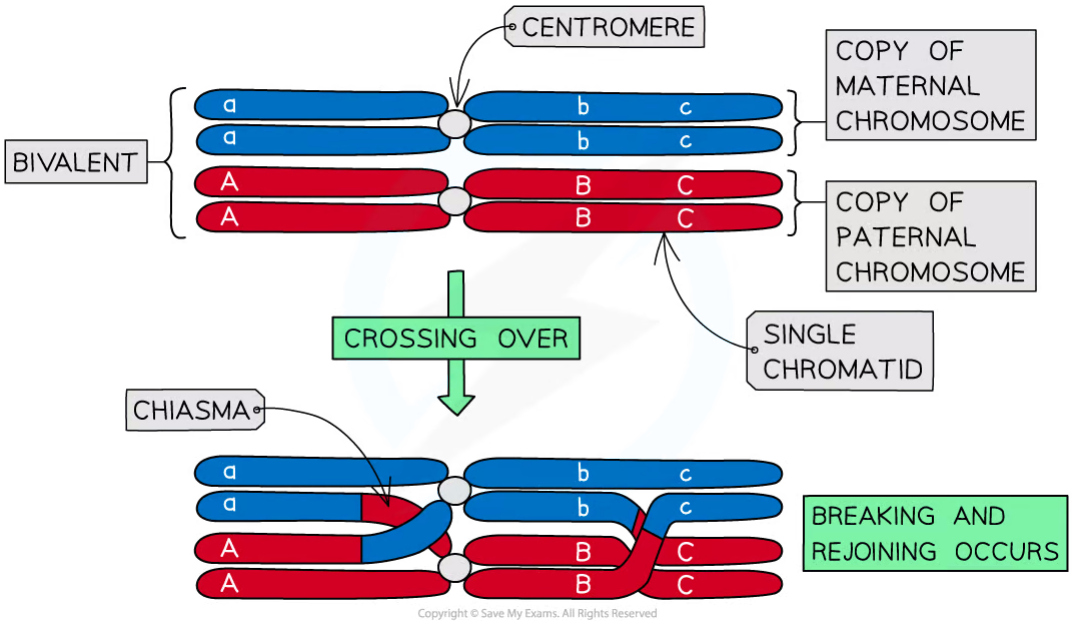
Non-sister chromatids originate from different parents' chromosomes and align during prophase 1

Chiasmata Formation

YOUR NOTES

Crossing over

- Crossing over is the process by which **non-sister chromatids exchange alleles**
- Process:
  - During prophase I, homologous chromosomes pair up and are in **very close proximity** to each other
  - The non-sister chromatids can **get entangled** and 'cross over' each other
  - The entanglement places **stress on the DNA molecules**
  - As a result of this molecular stress, a section of chromatid from one chromosome may **break and re-join** with the chromatid from the other chromosome
  - The breaking and re-joining is catalysed by **endonuclease** and **DNA ligase** enzymes respectively
- This swapping of alleles is significant as it can result in a new combination of alleles on the two chromosomes
- Any process that involves breaking and re-joining of DNA to create new combinations of genetic information is called recombination
  - DNA/chromosomes that have exchanged DNA in this way are referred to as **recombinant**
- When the DNA coils up, DNA strands at the crossing points remain attached to each other, so this causes the chromosome structure to change shape, developing an **X-shaped join**
- These crossing points are called chiasmata (singular: '**chiasma**')
- There is usually at least one, if not more, chiasma present in each bivalent during meiosis
- Crossing over does not just occur between non-sister chromatids that are immediately adjacent to each other
  - Crossing over can occur from one chromatid to either/both chromatids of the adjacent homologous pair



The formation of chiasmata (following synapsis)

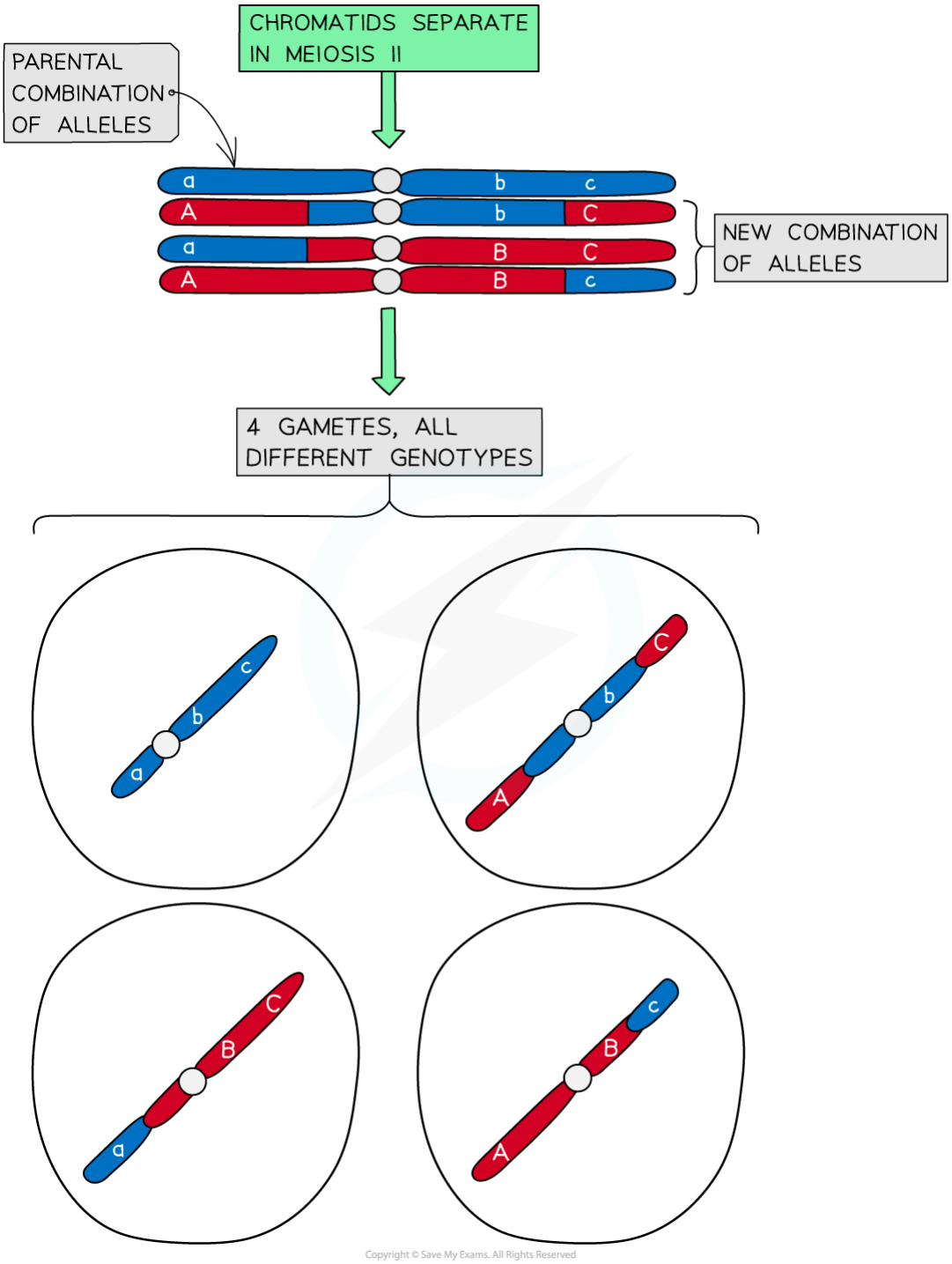
New Allele Combinations

YOUR NOTES

Crossing over produces new combinations of alleles on the chromosomes of the haploid cells

- So within a bivalent, or tetrad, there are 4 chromatids lying alongside each other and forming chiasmata
- Some of these chromatids will have **exchanged lengths of DNA** with each other at the chiasmata
- Each chromatid from a tetrad separates from the others during meiosis II
- Each chromatid goes on to form a haploid gamete
  - And so a different range of alleles will be carried in each gamete cell
  - This contributes greatly to intraspecific variation
- Back in prophase I, crossing over is more likely to occur further down the chromosome, away from the centromere
  - Because areas of DNA away from the centromere can flail around more, they are more likely to become entangled
  - Gene locus can therefore affect the genotype spread within a population, based on the likelihood of crossing over generating new allele combinations

YOUR NOTES



Distinct allele combinations appear in haploid gametes following crossing over

- Because of the random nature of how chromatids align and where they break, there is an almost **infinite range of combinations** of how DNA can recombine during crossing over
  - This explains that even in a very large population, **no two individuals will have exactly the same genotype** (except identical twins)

Exam Tip

Independent assortment can create a number of combinations that can be calculated. However, crossing over generates an incalculable amount of variation (we can assume it's infinite), just because of the random nature of where chiasmata can form.

YOUR NOTES

YOUR NOTES

Meiosis I

Homologous chromosomes separate in meiosis I

Prophase I

- DNA has already replicated and **condenses** and **becomes visible** as chromosomes
- Each chromosome consists of two sister chromatids joined together by a centromere
- The chromosomes are arranged side-by-side in homologous pairs
  - A pair of homologous chromosomes is called a **bivalent**
- As the homologous chromosomes are very close together the crossing over of non-sister chromatids may occur. The point at which the crossing over occurs is called the **chiasma** (chiasmata; plural)
- In this stage **centrioles migrate** to opposite poles and the spindle is formed
- The **nuclear envelope breaks down** and the **nucleolus disintegrates**

Metaphase I

- The **bivalents line up along the equator of the spindle**, with the spindle fibres attached to the centromeres
- The bivalents line up by **independent assortment** (random orientation)

Anaphase I

- The homologous pairs of chromosomes are separated **as microtubules pull whole chromosomes to opposite ends** of the spindle
- The centromeres do not split

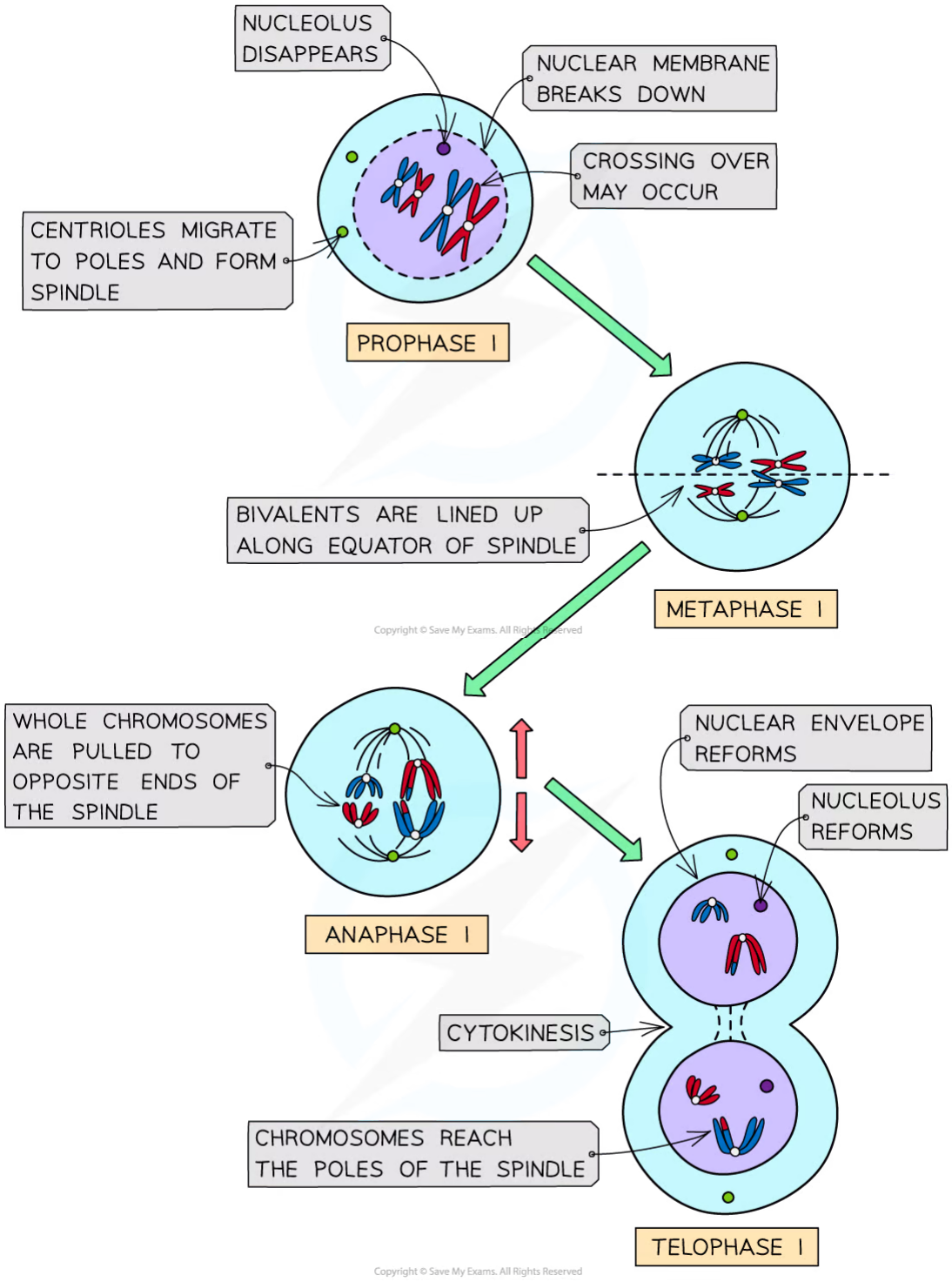
Telophase I

- The chromosomes arrive at opposite poles
- Spindle fibres start to break down
- **Nuclear envelopes form** around the two groups of chromosomes and nucleoli reform
- Some plant cells go straight into meiosis II without reformation of the nucleus in telophase I

Meiosis I is reduction division

- Meiosis I is referred to as **reduction division** because homologous chromosomes separate and move to opposite poles of the cell.
- Therefore, the number of chromosomes per cell is **reduced** by a factor of 2

YOUR NOTES



The different stages of meiosis I in an animal cell

Exam 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.

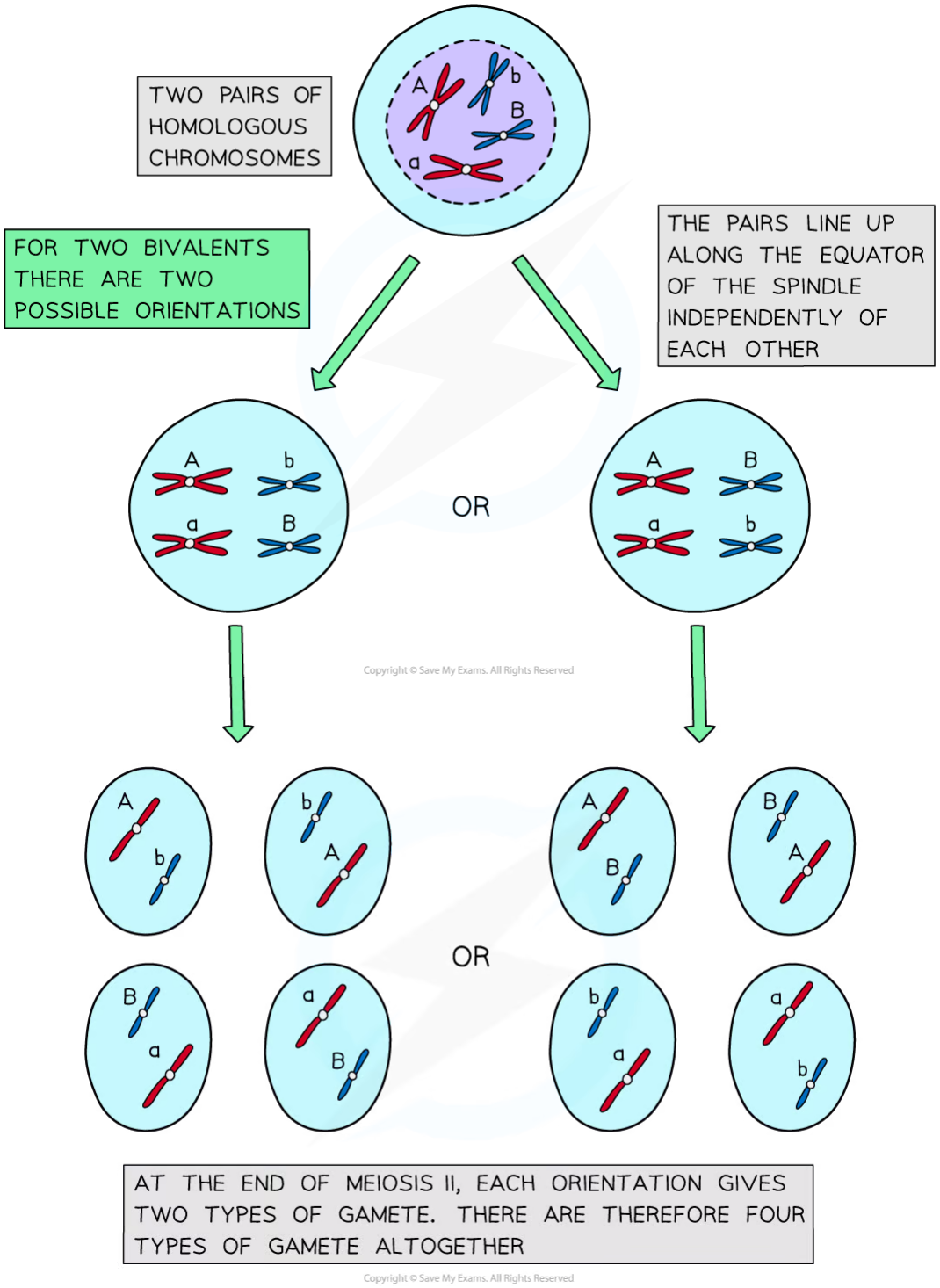
YOUR NOTES

Independent Assortment

YOUR NOTES

- During metaphase I, an event occurs that is an important source of genetic variation in the gametes formed by meiosis
- Independent assortment is the **production of different combinations of alleles** in gamete cells due to the random alignment of homologous pairs along the equator of the spindle during metaphase I
  - This random alignment is sometimes referred to as **random orientation**
- In prophase I homologous chromosomes pair up and in metaphase I they 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 during anaphase
- 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 **8 388 608 different combinations**
  - This may seem like a lot of combinations, but by contrast, **crossing over** introduces an almost infinite amount of variation

YOUR NOTES



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

Explaining Deviations from Mendelian Ratios

YOUR NOTES

NOS: Careful observation and record keeping turned up anomalous data that Mendel's law of independent assortment could not account for

- Gregor Mendel (of pea plant fame) devised the **law of independent assortment** in 1866
- In this, he states that characteristics are inherited **completely independently** of others
- This was refined by later scientists to state that the allele that gets sorted into a particular gamete cell **is not influenced** by the allele received for another gene
- In many cases, this holds true, and can be observed in **Mendelian ratios** of offspring in certain crossing experiments
  - Mendel observed a 9:3:3:1 ratio in many dihybrid crosses of sweet pea plants
- As a result, **Mendel's findings were not challenged** until the early 20th century, when Bateson and Punnett found **seemingly anomalous** ratios of offspring
- For which they could offer no explanation
- These became named as **non-Mendelian ratios** because they did not follow the pattern as predicted by Mendel

Bateson and Punnetts' experiment (1905)

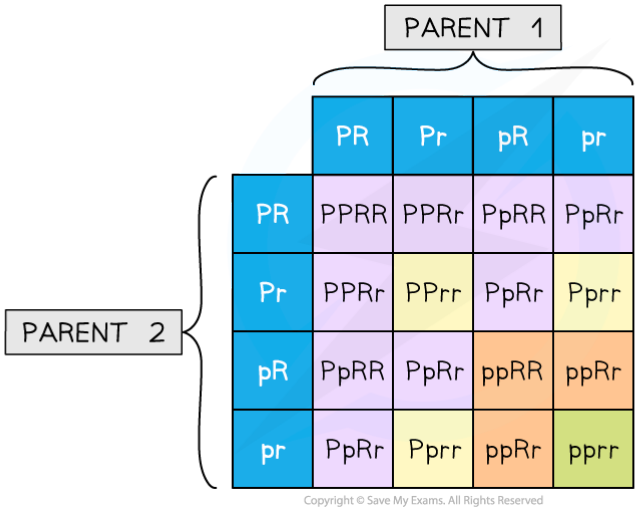
- Also working with sweet peas, **two pairs of alleles** were identified (Dominant alleles in capital letters)
  - Flower colour:
    - ***P*** = purple, ***p*** = red
  - The shape of pollen grains:
    - ***R*** = long, ***r*** = round
- Their first cross involved pure-bred purple-flowered plants with long pollen grains (***PPRR***) with pure-bred red-flowered plants with round pollen grains (***pprr***)
- As expected, this cross resulted in 100% purple-flowered plants with long pollen grains, with the double-heterozygous genotype ***PpRr*** in the F_1 generation
- However, when individuals from the F_1 generation were crossed with each other, Bateson and Punnett **would have expected a 9:3:3:1 mix of phenotypes**, in line with Mendel's law of independent assortment

Modelling the expected ratios using a Punnet square

Parental phenotypes:	purple flowers, long pollen grains	x	purple flowers, long pollen grains
Parental genotypes:	***PpRr***	x	***PpRr***
Parental gametes:	***PR*** or ***Pr*** or ***pR*** or ***pr***	x	***PR*** or ***Pr*** or ***pR*** or ***pr***

Dihybrid Cross Punnett Square Table

YOUR NOTES



- Expected phenotype ratios for the second generation:
  - 9 purple flowers, long pollen grains
  - 3 purple flowers, round pollen grains
  - 3 red flowers, long pollen grains
  - 1 red flowers, round pollen grains

Actual Results from Bateson and Punnetts' Experiment

- In the F_2 generation of offspring, there were...
  - **Very many** of the grandparent phenotype (purple-flowered, long pollen grains) produced
  - **Many** of the grandparent phenotype (red-flowered, round pollen grains) produced
  - **Very few (but not zero)** other phenotypes (purple/round or red/long) produced
- This **appeared as an anomaly** to Bateson and Punnett, though they did not find an explanation

Thomas Hunt Morgan developed the notion of linked genes to account for the anomalies

- Later in the 20th century (approx. 1910–1940), an American biologist, **Thomas Hunt Morgan**, put forward an explanation for the results previously observed by Bateson and Punnett
- From his findings, he devised the theory of linkage
- Morgan worked on **fruit flies** (*Drosophila melanogaster*) thanks to their ability to reproduce **quickly**, in **large numbers**, in a small physical space and with **observable heritable characteristics** like eye colour and wing shape
- In fact, Morgan bred them selectively for many years to develop a range of phenotypes by natural mutation
- Morgan's work identified sex-linked characteristics from genes carried on the X or Y chromosomes that determine gender
- This work led Morgan to develop the idea of autosomal linkage
- This is where **unexpected patterns of inheritance** are caused by separate alleles being inherited **together**, from the same chromosome (an autosome)
- Morgan then elaborated his work by developing the theory of **crossing over**

- As a way of accounting for unexpected (recombinant) genotypes

YOUR NOTES

Exam Tip

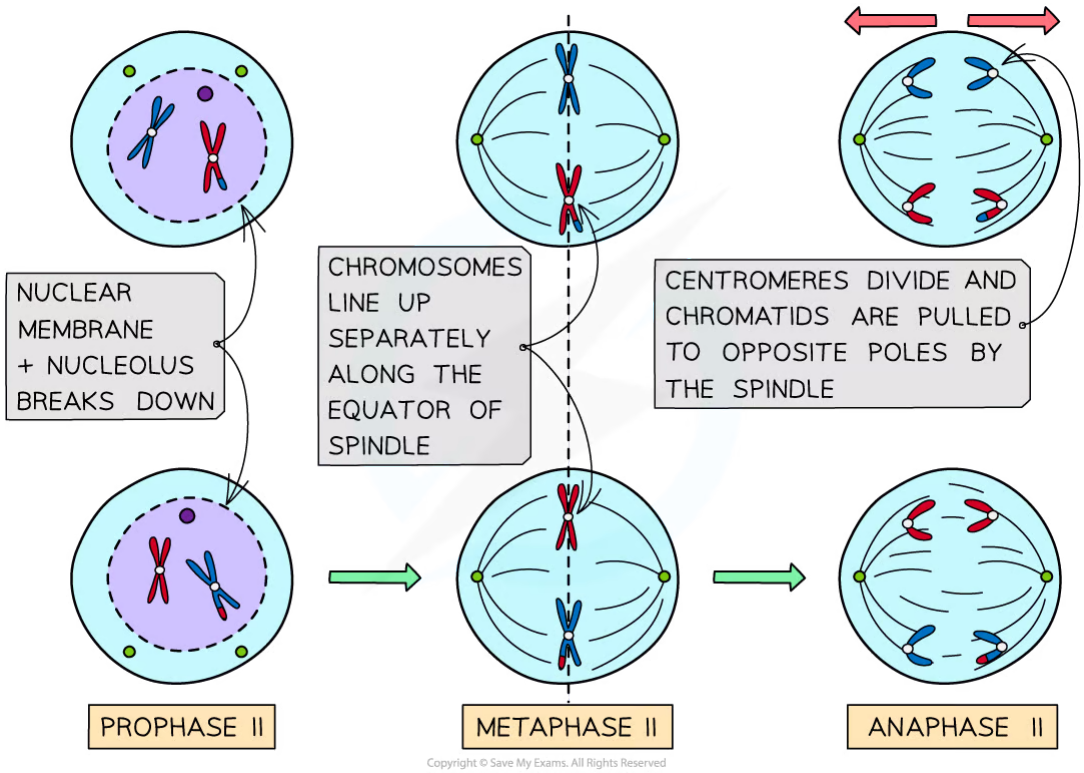
Several sources of genetic variation have been outlined above. It is also worth remembering that genetic variation can occur on an even smaller scale than chromosomes. Mutations can occur within genes. A random mutation that takes place during DNA replication can lead to the production of new alleles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

YOUR NOTES

Meiosis II

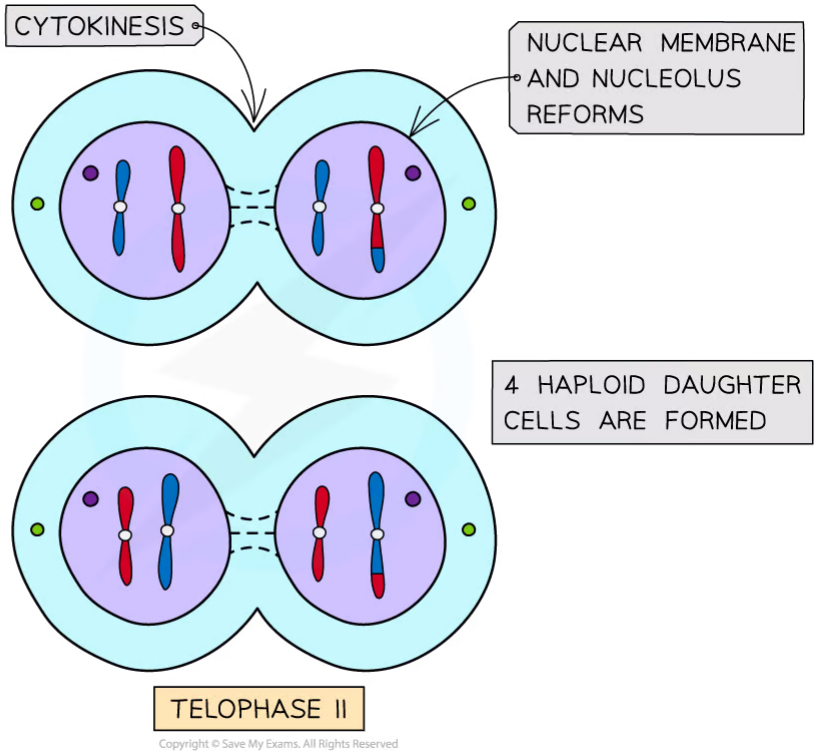
Second division of Meiosis : Meiosis II

- There is **no interphase between meiosis I and meiosis II** so the DNA is not replicated
- The second division of meiosis is almost identical to the stages of mitosis
- Prophase II
  - The **nuclear envelope breaks down** and **chromosomes condense**
  - A **spindle forms at a right angle to the old one**
- Metaphase II
  - **Chromosomes** line up in a **single file along the equator** of the spindle
- Anaphase II
  - Centromeres divide and individual **chromatids are pulled to opposite poles**
  - Sister chromatids separate in meiosis (anaphase) II
    - However, they are likely to be **non-identical sister chromatids** at this stage due to crossing over having happened in prophase I
  - This creates **four groups of chromosomes** that have half the number of chromosomes compared to the original parent cell
- Telophase II
  - **Nuclear membranes form** around each group of chromosomes
- Cytokinesis
  - **Cytoplasm divides** as new cell surface membranes are formed **creating four haploid cells**



Prophase II, Metaphase II and Anaphase II in Meiosis II of an animal cell

YOUR NOTES



Telophase II and cytokinesis in Meiosis II of an animal cell

Exam Tip

Because of the many similarities between mitosis and meiosis II, you are more likely to get a detailed question on meiosis I than meiosis II and the sources of variation that occur in meiosis I. Revise them both though!

YOUR NOTES

Drawing Chiasmata

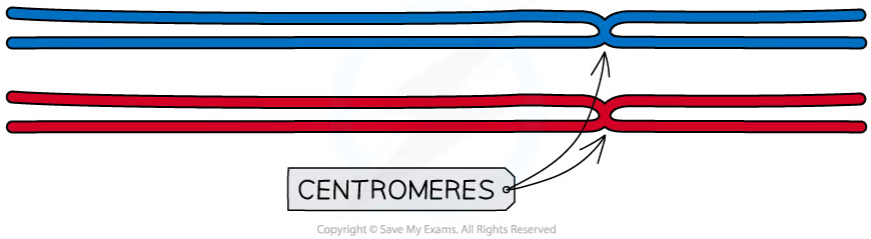
Skill: Drawing diagrams to show chiasmata formed by crossing over

Drawing tips

- Use two coloured pens/pencils to show chromosomes/chromatids of maternal or paternal origin
  - One chromosome of each colour makes a homologous pair
  - Blue and red are conventionally used for this purpose, but any colour choices that show good contrast are acceptable
  - Draw each homologous chromatid as a long line

Stage 1: Synapsis

- All 4 chromatids of a pair of homologous chromosomes align closely together
- Draw this as 4 lines in close proximity, 2 red and 2 blue
  - Remember to include the centromeres

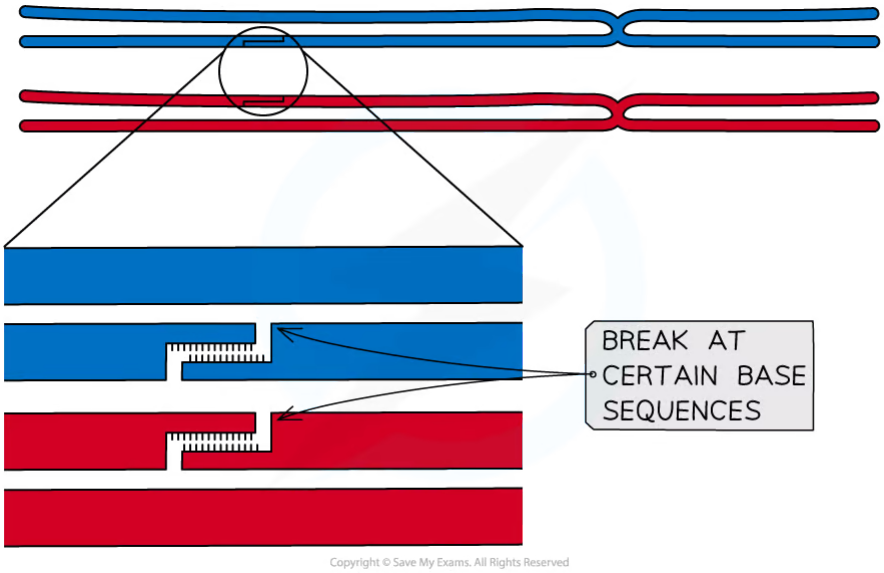


Stage 1 : Synapsis. A bivalent (tetrad) forms from two homologous chromosomes. There are 4 chromatids aligned against each other

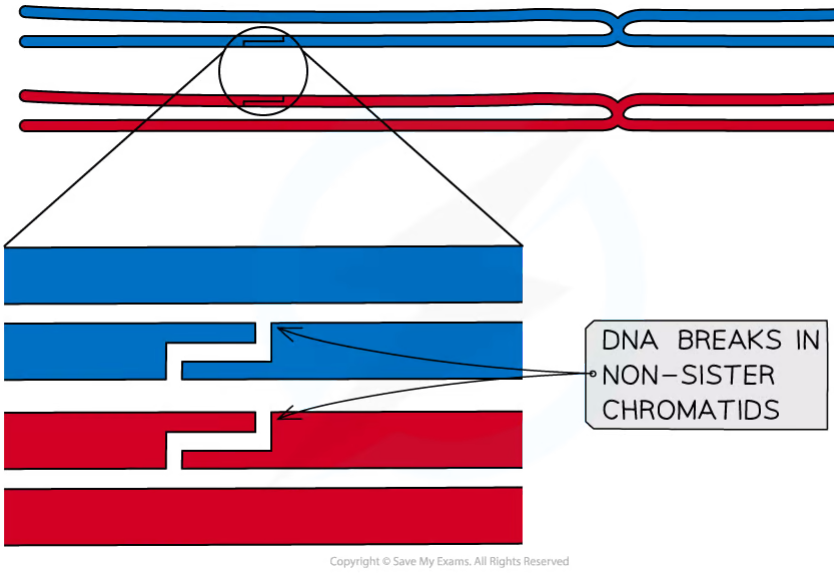
Stage 2: Cuts occur in the DNA of non-sister chromatids

- During **coiling and shortening of DNA** in prophase 1, the DNA is stressed/placed under tension
- This causes a **cut in the DNA** of one of the chromatids, catalysed by **endonuclease** enzymes
  - In fact, **many cuts occur simultaneously** within the same bivalent
- One such cut is shown below
- The adjacent non-sister chromatid also breaks **at the same point** as it has the same base sequence at the point of breakage
  - And is cut by the same endonuclease enzyme

YOUR NOTES



Stage 2: Cuts occur in the DNA of non-sister chromatids



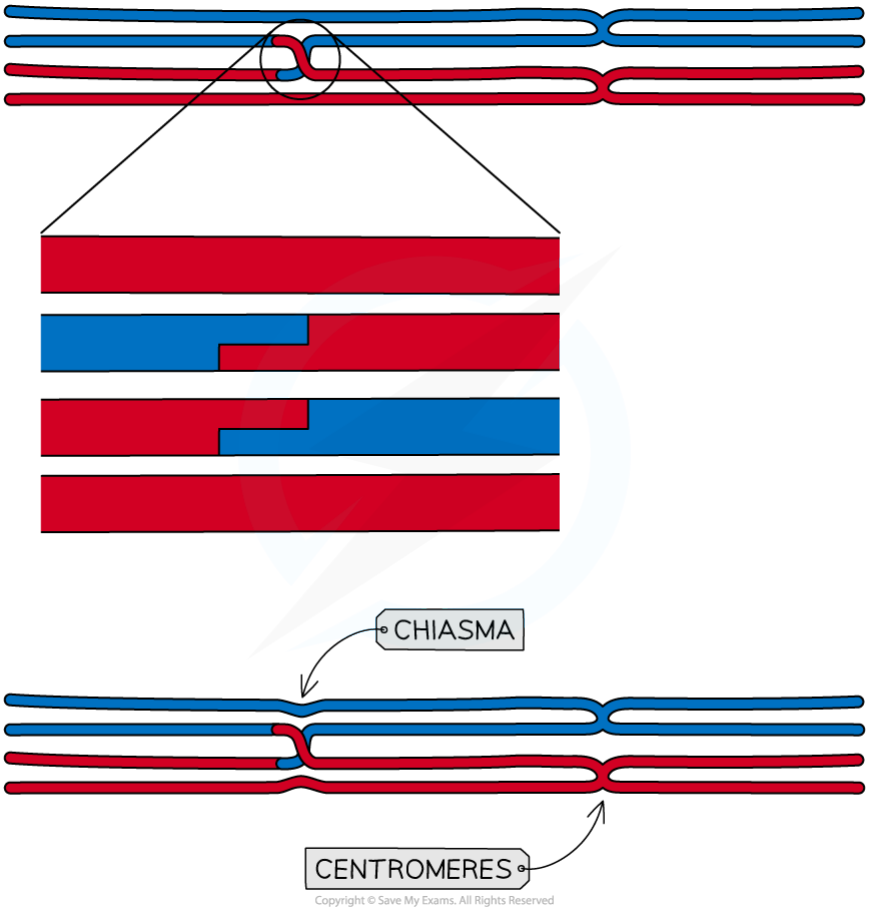
Stage 2: Cuts occur in the DNA of non-sister chromatids

Stage 3: Formation of Chiasmata

- There are **loose, cut ends of DNA** within the bivalent with short sections of exposed, unpaired bases
- These bases **re-form hydrogen bonds** to complementary bases quickly, however,
- They can base-pair to cut ends from a **different chromatid**
- This can occur with a non-sister chromatid because the non-sister chromatid will have a very similar, **almost identical sequence of bases**
- This is how crossing-over leads to **swapping of alleles** between non-sister chromatids
- When the chromosomes condense and shorten again, the chiasmata continue to hold non-sister chromatids together

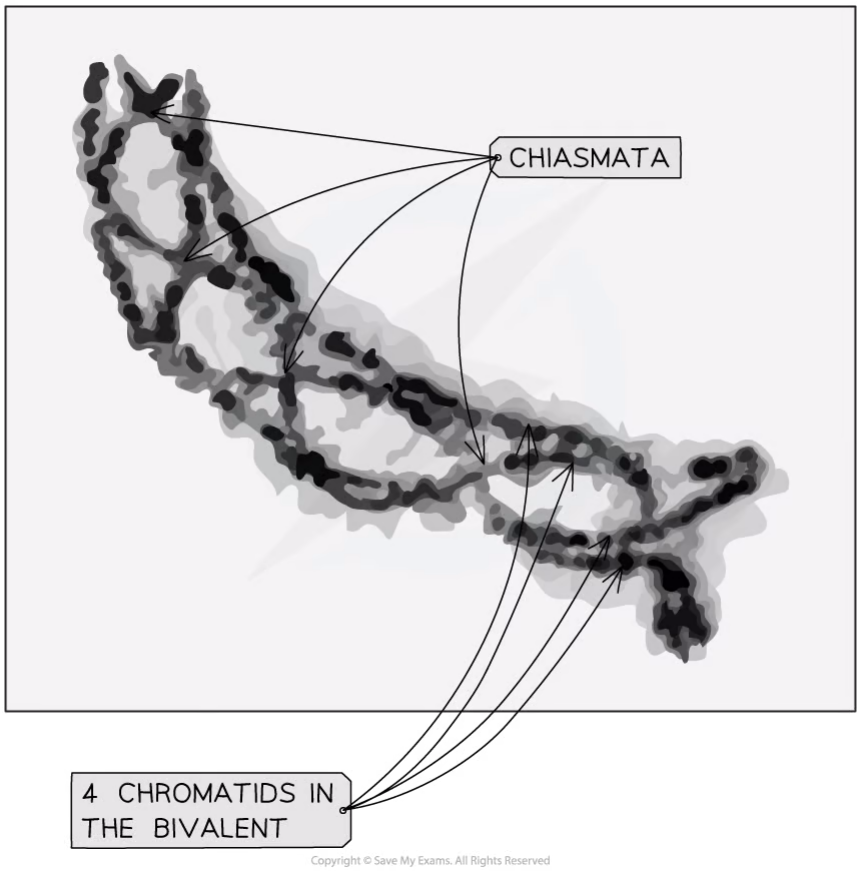
YOUR NOTES

- This causes the overall chromosome shape to feature **X-shapes** at the chiasmata, viewable under an **electron microscope**



Stage 3: Appearance of the recombinant bivalent, with chiasma showing as an X-shape

YOUR NOTES



Electron micrograph of a bivalent in prophase 1, showing multiple chiasmata

YOUR NOTES

10.2 Inheritance

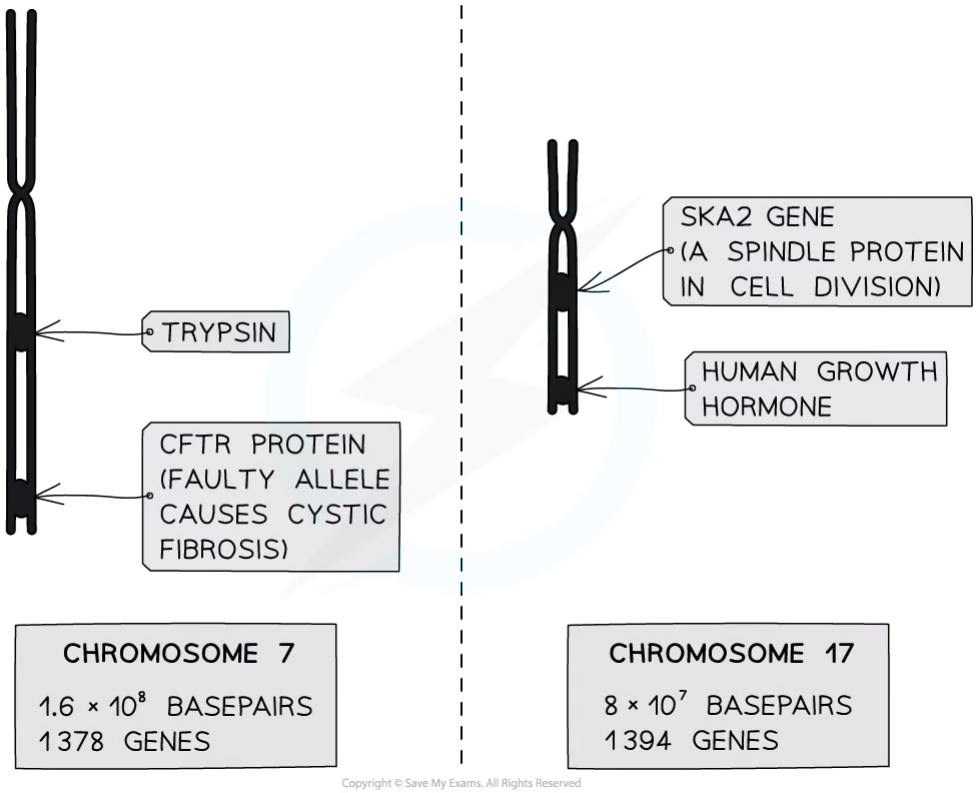
10.2.1 Unlinked Genes

Independent Assortment & Segregation

Unlinked genes segregate independently as a result of meiosis

- **Unlinked genes** are genes that an organism carries on **separate chromosomes**
  - Not on homologous copies of the same chromosome
- An example of a pair of unlinked genes in fruit flies (*Drosophila melanogaster*) is
  - The gene for curly wings on **chromosome 2**, and
  - The gene for mahogany eyes on **chromosome 3**
- An example of a pair of unlinked genes in humans is
  - The gene for trypsin (a stomach enzyme) on chromosome **7**, **and**
  - The gene for human growth hormone on chromosome **17**
- **Assortment** of chromosomes refers to their alignment in metaphase I of meiosis
  - Each bivalent assorts (aligns) itself independently of all the others
- **Segregation** of chromosomes (ie. how they get separated) is governed by their pattern of assortment
  - Segregation just refers to **which pole of the cell** the whole chromosomes are pulled to in anaphase I
  - Segregation determines **which combinations of alleles** end up in which gamete cells by the end of meiosis II
- By contrast, linked genes (on the same chromosome) tend to be **inherited together**

YOUR NOTES



The loci of selected genes in the human genome

Trypsin and CFTR are linked genes (both on the same chromosome);

Human Growth Hormone and trypsin are unlinked genes (both on different chromosomes)

Punnett Squares for Dihybrid Traits

YOUR NOTES

- **Mono**hybrid crosses look at how the alleles of **one** gene transfer across generations
- **Di**hybrid crosses look at how the alleles of **two** genes transfer across generations
  - ie. dihybrid crosses can be used to show the **inheritance** of **two completely different characteristics** in an individual
- The genetic diagrams for both types of cross are very similar
- For dihybrid crosses, there are several more genotypes and phenotypes involved
- When writing out the different genotypes, write the **two alleles for one gene**, followed immediately by the **two alleles for the other gene**.
- Do not mix up the alleles from the different genes
  - For example, if there was a gene with alleles ***Y*** and ***y*** and another gene with alleles ***G*** and ***g*** an example genotype for an individual would be ***YyGg***
- Alleles are usually shown side by side in dihybrid crosses e.g. ***TtBb***

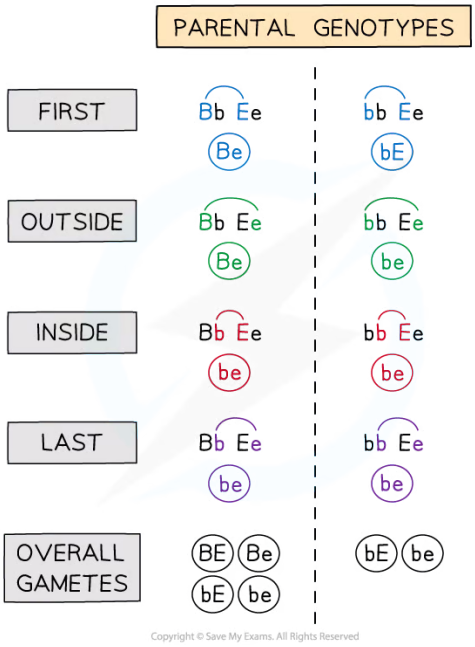
YOUR NOTES

Worked Example

Worked example 1: Dihybrid genetic diagram

- Horses have a single gene for coat colour that has two alleles:
  - ***B***, a dominant allele produces a black coat
  - ***b***, a recessive allele produces a chestnut coat
- Horses also have a single gene for eye colour
  - ***E***, a dominant allele produces brown eyes
  - ***e***, a recessive allele produces blue eyes
- Each of these genes (consisting of a pair of alleles) are **inherited independently** of one another because the two genes are located on different non-homologous chromosomes
  - Such characteristics are said to be unlinked
- In this example, a horse that is heterozygous for both genes has been crossed with a horse that is homozygous for one gene and heterozygous for the other

Parental phenotypes:	black coat, brown eyes	x	chestnut coat, brown eyes
Parental genotypes:	***BbEe***	x	***bbEe***
Parental gametes:	***BE*** or ***Be*** or ***bE*** or ***be***	x	***bE*** or ***be***



Determining the Alleles Carried by Gametes Based on the Parental Genotypes Using the FOIL (First, Outside, Inside, Last) Method

Dihybrid Cross Punnett Square Table

YOUR NOTES

		Gametes from Parent Two	
		bE	be
Gametes from Parent One	BE	BbEE / black coat, brown eyes	BbEe / black coat, brown eyes
	Be	BbEe / black coat, brown eyes	Bbee / black coat, blue eyes
	bE	bbEE / chestnut coat, brown eyes	bbEe / chestnut coat, brown eyes
	be	bbEe / chestnut coat, brown eyes	bbee / chestnut coat, blue eyes

- Predicted ratio of phenotypes in offspring =
  - **3** black coat, brown eyes :
  - **3** chestnut coat, brown eyes :
  - **1** black coat, blue eyes :
  - **1** chestnut coat, blue eyes
- Predicted ratio of genotypes in offspring = **3** ***BbEE*** **: 3** ***bbEE*** **: 1** ***Bbee*** **: 1** ***bbee***

Worked Example

Worked example 2: Dihybrid genetic diagram

In a separate cross to that shown in Worked Example 1, a horse that is heterozygous for both genes has been crossed with another horse that is heterozygous for both genes.

Parental phenotypes:	black coat, brown eyes	x	black coat, brown eyes
Parental genotypes:	***BbEe***	x	***BbEe***
Parental gametes:	***BE*** or ***Be*** or ***bE*** or ***be***	x	***BE*** or ***Be*** or ***bE*** or ***be***

Dihybrid Cross Punnett Square Table 2

YOUR NOTES

		Gametes from Parent Two			
		BE	Be	bE	be
Gametes from Parent One	BE	BBEE / black coat, brown eyes	BBEe / black coat, brown eyes	BbEE / black coat, brown eyes	BbEe / black coat, brown eyes
	Be	BBEe / black coat, brown eyes	BBee / black coat, blue eyes	BbEe / black coat, brown eyes	Bbee / black coat, blue eyes
	bE	BbEE / black coat, brown eyes	BbEe / black coat, brown eyes	bbEE / chestnut coat, brown eyes	bbEe / chestnut coat, brown eyes
	be	BbEe / black coat, brown eyes	Bbee / black coat, blue eyes	bbEe / chestnut coat, brown eyes	bbee / chestnut coat, blue eyes

- Predicted ratio of phenotypes in offspring =
  - **9** black coat, brown eyes :
  - **3** chestnut coat, brown eyes :
  - **3** black coat, blue eyes :
  - **1** chestnut coat, blue eyes

Exam Tip

For the double-heterozygous cross for unlinked genes above, you're expected to remember the phenotypic ratio 9:3:3:1. You won't need to remember the ratio of the genotypes but this can be worked out from a Punnett square like the one above.

YOUR NOTES

Predicting Phenotypic and Genotypic Ratios

Worked Example

Fruit flies (*Drosophila melanogaster*) were crossed in a laboratory study looking for inheritance patterns for two characteristics, wing length and body colour. We can assume that these characteristics are unlinked.

The alleles for these characteristics are as follows:

V = long wings

v = short (vestigial) wings

B = brown body colour

b = black body colour

A black-bodied, heterozygous long-winged fly was crossed with short-winged, homozygous brown-bodied flies. Predict the phenotype ratio of their offspring.

Step 1: Write out the parental genotypes

The first parent is black-bodied and heterozygous long-winged. To be black-bodied it must have the genotype ***bb***. Heterozygous long-winged has the genotype ***Vv***
The second parent is short-winged and homozygous brown-bodied. To be short-winged it must have the genotype ***vv***. Homozygous brown-bodied has the genotype ***BB***

Vvbb × ***vvBB***

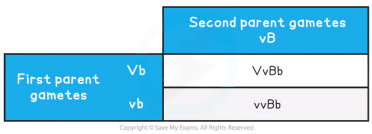
Step 2: Identify the gamete genotypes that each parent could produce

These are the allele combinations that each parent can produce in meiosis

Vb ***vb*** × ***vB***

Step 3: Complete a Punnett square to show the genotypes of the offspring

Punnett Square showing Genotypes of the Offspring



Step 4: Identify the phenotypes of the offspring

VvBb = long-winged, brown-bodied
vvBb = short-winged, brown bodied

Conclusion: The offspring would be 100% brown-bodied and 1:1 long to short-winged

Test crosses

YOUR NOTES

- A test cross can be used to **deduce the genotype**
- The individual in question is crossed with an individual that is expressing the **recessive phenotype**
- This is because an individual with a recessive phenotype has a known genotype
- The resulting phenotypes of the offspring provide sufficient information to suggest the genotype of the unknown individual
- For a monohybrid test cross:
  - If **no** offspring exhibit the recessive phenotype then the unknown genotype is **homozygous dominant**
  - If **at least one** of the offspring exhibit the recessive phenotype then the unknown genotype is **heterozygous**
- For a dihybrid test cross:
  - If no offspring exhibit the recessive phenotype for either gene then the unknown genotype is **homozygous dominant** for both genes
  - If at least one of the offspring exhibit the recessive phenotype for one gene but not the other, then the unknown genotype is **heterozygous for one gene and homozygous dominant for the other**
  - If at least one of the offspring exhibit the recessive phenotype for both genes then the unknown genotype is **heterozygous for both** genes

Worked Example

Worked example: Test crosses

- Rabbits have a single gene for ear length that has two alleles:
  - ***D***, a dominant allele that produces long ears
  - ***d***, a recessive allele that produces shorter ears
- A breeder has a rabbit called Floppy that has long ears and they want to know the genotype of the rabbit
  - There are two possibilities: ***DD*** or ***Dd***
- The breeder crosses the long-eared rabbit with a short-eared rabbit
  - A rabbit displaying the recessive short ear phenotype has to have the genotype ***dd***

Test Cross Possibility Table

		Known gametes	
		d	d
Possible gametes Option 1	D	Dd / long ears	Dd / long ears
	D	Dd / long ears	Dd / long ears

- The predicted ratio of phenotypes of offspring – 1 long ears
- The predicted ratio of genotypes of offspring – 1 Dd

Test Cross Possibility Two Table

YOUR NOTES

		Known gametes	
		d	d
Possible gametes Option 2	D	Dd / long ears	Dd / long ears
	d	dd / short ears	dd / short ears

- Predicted ratio of phenotypes of offspring – 1 long ears : 1 short ears
- Predicted ratio of genotypes of offspring – 1 *Dd* : 1 *dd*
- The breeder identifies the different phenotypes present in the offspring
- There is at least one offspring with the short ear phenotype
- This tells the breeder that their rabbit Floppy has the genotype ***Dd***
- If Floppy was genotype ***DD*** none of the offspring would have short ears

Worked Example

Worked example: Hard Question

A farmer wishes to maximise his yield of soybean oil from his crop. Oil is extracted from the seeds which typically measure 6–12 mm in diameter. In one species of soybean, Glycine max, two characteristics are governed by the following pairs of **unlinked** alleles.

H = high oil content in the seeds; ***h*** = low oil content in the seeds

E = four seeds in a pod; ***e*** = two seeds in a pod

The farmer crossed two soybean plants, both with high oil content and four seeds per pod. This cross resulted in 381 offspring plants being produced. The 381 F_1 offspring had a phenotypic spread as shown in the table below.

Phenotype of F_1 generation		Numbers of plants in F_1 generation
Oil Content	Number of Seeds per Pod	
High	4	215
High	2	73
Low	4	70
Low	2	23
		381

Use the data in the table to deduce the genotypes of the parent plants in this cross.

Step 1: Write out the possible parental genotypes

Both parents were high-oil content with 4 seeds per pod, which are the dominant traits for each pair of alleles. Neither parent can be homozygous recessive for either oil content or the number of seeds.

Possible allele combinations for oil content: ***HH***, ***Hh***

Possible allele combinations for 4 seeds per pod: ***EE***, ***Ee***

YOUR NOTES

Therefore, the possible parental genotypes were: ***HHEE***, ***HHEe***, ***HhEE***, ***HhEe***

Step 2: Examine phenotype ratios

The smallest number of F_1 offspring was 23 for low oil content, 2 seeds. These must have the homozygous recessive genotype, ***hhee***.

The ratios of other phenotypes, relative to the homozygous recessive phenotype, were:
low oil, 4 seeds : ***hhee*** = 70:23 = 3.04 : 1 ≅ 3:1
high oil, 2 seeds : ***hhee*** = 73:23 = 3.17 : 1 ≅ 3:1
high oil, 4 seeds : ***hhee*** = 215:23 = 9.3 : 1 ≅ 9:1

Step 3: Apply Mendel's Law of independent assortment for linked genes

The phenotypes of the offspring display an approximate 9:3:3:1 ratio. This characterises a cross between two double-heterozygous parents where the genes are unlinked, as in this case

Conclusion: Both parents' genotype was ***HhEe*** / double heterozygous

Exam Tip

Make sure before you start a test cross you think about the following: how many genes are there, how many alleles of each gene are there, which is the dominant allele, what type of dominance is it and is there linkage or codominance between genes?

Even though we learn about fruit flies (*Drosophila melanogaster*) being studied in detail which led to the discovery of gene linkage, many of their characteristics are unlinked and can be studied on a large scale in this manner.

YOUR NOTES

Exceptions to Mendel's Rules

NOS: Mendel used observations of the natural world to find and explain patterns and trends

- Since Mendel, scientists have looked for discrepancies and asked questions based on further observations to show exceptions to the rules. For example, Morgan discovered non-Mendelian ratios in his experiments with *Drosophila*
- When looking at dihybrid crosses (crosses with two pairs of observable characteristics), Mendel explained his experimental data with his **law of independent assortment**
- That individual characteristics are **inherited completely independently** of each other
- In many cases, **this is correct**
- The significance of Mendel's work went **largely unnoticed for decades**, until after his death in 1884
- Scientists in the 1890s and early 1900s picked up his experimental findings and **replicated them**
- The **large number of trials** that Mendel undertook highlighted patterns/trends in the inheritance of certain factors (what are now known as genes)
- However, **discrepancies were noticed** when scientists replicated Mendel's experiments
  - And also when experiments were undertaken on the inheritance of certain genes in **other organisms**
- Many of their dihybrid crosses replicated the **9:3:3:1 pattern of phenotypes** that Mendel first observed in his work
- **William Bateson** and **Reginald Punnett**, two Cambridge University geneticists (in collaboration with a third Cambridge biologist, **Edith Saunders**), replicated Mendel's findings, again in experiments with sweet peas, however
- They discovered some **apparently anomalous results** in certain cases, in which phenotype ratios **did not follow** the classical 9:3:3:1 pattern
- Many scientists **would have dismissed non-conforming** results as mere anomalies, however Bateson, Punnett and Saunders chose to search for an explanation
- Punnett's quote from one his **laboratory notebooks** sums up their approach:
  - "*Treasure your exceptions! When there are none, the work gets so dull that no one cares to carry it further.*"
- Bateson and Punnett performed further work, mainly on crossings of sweet peas and crossings of chickens, but were **unable to offer a robust explanation** for certain unpredictable phenotype ratios in their crosses

Gene Linkage

YOUR NOTES

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
- If genes are on the sex chromosome, they are said to be **sex-linked**
  - Sex-linked genes have characteristics that generally **only affect one gender** of a species
  - These genes are usually **on the X chromosome** because the Y chromosome contains very few genes
  - In humans, **colour-blindness** and **haemophilia** are notable examples of genetic conditions that only affect males
- Linked genes located on the chromosomes 1–22, or any chromosome that is not a sex chromosome (called autosomes) are said to be examples of **autosomal linkage**
- The likelihood of genes being inherited together, or the extent to which they are linked, is measured in units called **centimorgans**, in honour of Thomas Hunt Morgan's work

Notation for link genes

- When writing linked genotypes it can be easier to keep the linked alleles within a bracket
  - For example, an individual has the genotype ***FFGG***. However, if there is linkage between the two genes, it would be written as (***FG***)(***FG***)
- Another commonly-used way of denoting linked alleles is to link them with a line. So, for example, linkage between genes ***F*** and ***G*** might be shown as

F ***G***

_____

_____

f ***g***

- Remember to distinguish between sex linkage and autosomal linkage. The explanation of non-Mendelian ratios falls into the domain of autosomal linkage for IB

Autosomal linkage

- Dihybrid crosses and their predictions rely on the assumption that the genes being investigated behave **independently of one another** during meiosis
- However, **not all genes assort independently** during meiosis
- Some genes which are located on the **same chromosome** display autosomal **linkage** and **stay together in the original parental combination**
- Linkage between genes affects how parental alleles are passed onto offspring through the gametes

Identifying autosomal linkage from phenotypic ratios

- In the following **theoretical example**, a dihybrid cross is used to predict the inheritance of **two different characteristics** in a species of newt
  - The genes are for **tail length** and **scale colour**
- The gene for tail length has two alleles:
  - Dominant allele **T** produces a normal length tail
  - Recessive allele **t** produces a shorter length tail

YOUR NOTES

- The gene for scale colour has two alleles:
  - Dominant allele **G** produces green scales
  - Recessive allele **g** produces white scales

Without linkage

- The outcomes for this dihybrid cross if the genes are **unlinked** are as follows

Dihybrid Cross without Linkage Punnett Square Table

		Gametes from Parent Two
		tg
Gametes from Parent One	TG	TtGg / normal tail, green scales
	Tg	Ttgg / normal tail, white scales
	tG	ttGg / short tail, green scales
	tg	ttgg / short tail, white scales

- Predicted ratio of phenotypes in offspring =
  - 1 normal tail, green scales : 1 normal tail, white scales : 1 short tail, green scales : 1 short tail, white scales
- Predicted ratio of genotypes in offspring =
  - 1 TtGg : 1 Ttgg : 1 ttGg : 1 ttgg

With linkage

- However, if the **same dihybrid cross** is carried out but this time the genes are **linked**, we get a **different phenotypic ratio**
  - There would be a **1** : **1** phenotypic ratio (**1** normal tail, green scales : **1** short tail, white scales)
  - This change in the phenotypic ratio occurs because the genes are located on the **same chromosome**
  - The **unexpected phenotypic ratio**, therefore, shows us that the genes are **linked**
- The explanation for this new phenotypic ratio is given in the worked example below:

YOUR NOTES

Worked Example

Worked example: Explaining autosomal linkage

- In reality, the genes for tail length and scale colour in this particular species of newt show autosomal linkage

  Parental phenotypes: normal tail, green scales x short tail, white scales

  Parental genotypes: **(TG)(tg)** **(tg)(tg)**

  Parental gametes: **(TG)** or **(tg)** **(tg)**

Dihybrid Cross with Linkage Punnett Square Table

		Gametes from Parent Two
		(tg)
Gametes from Parent One	(TG)	(TG)(tg) / normal tail, green scales
	(tg)	(tg)(tg) / short tail, white scales

- Predicted ratio of genotypes in offspring =
  - **1** (TG)(tg) : **1** (tg)(tg)
- Predicted ratio of phenotypes in offspring =
  - **1** normal tail, green scales : **1** short tail, white scales

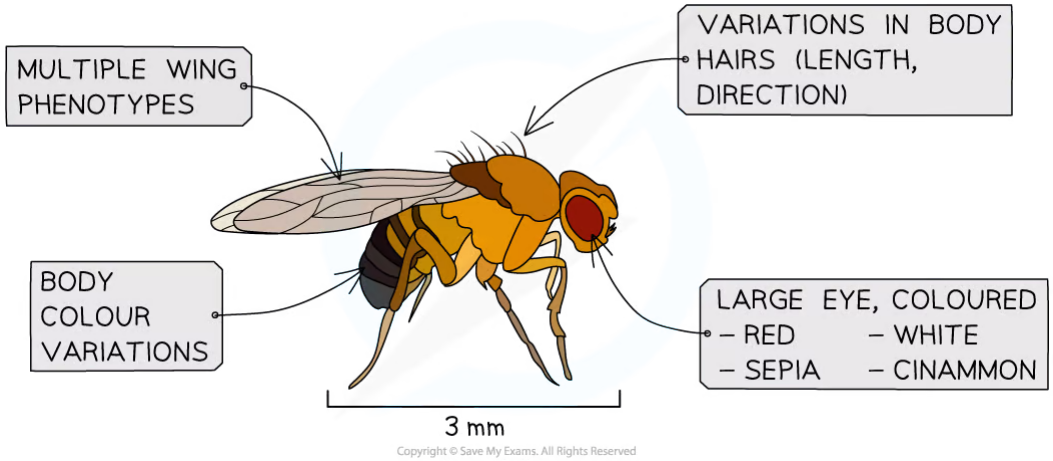
Exam Tip

When you are working through different genetics questions you may notice that test crosses involving autosomal linkage predict solely **parental type** offspring (offspring that have the same combination of characteristics as their parents). However in reality **recombinant** offspring (offspring that have a different combination of characteristics to their parents) are often produced. This is due to the **crossing over** that occurs during meiosis. The crossing over and exchanging of genetic material **breaks the linkage** between the genes and recombines the characteristics of the parents. So if a question comes along that asks you why recombinant offspring are present you now know why!

YOUR NOTES

Non-Mendelian Ratios in Drosophila

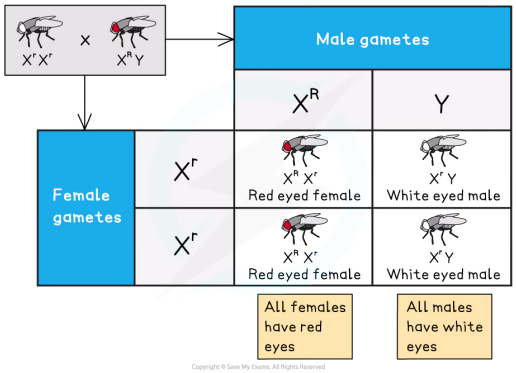
- In the way that Bateson, Punnett and Saunders developed and refined Mendel's findings, Thomas Hunt Morgan further refined genetic theory
  - His work was awarded the Nobel Prize in 1933



A fruit fly* (Drosophila melanogaster) *with some of the phenotypic variations observed in Thomas Hunt Morgan's work

Sex linkage

- Working in the USA in the early 20th century, he 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 phenotypic distribution**
  - 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. X-linked)



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

Autosomal linkage

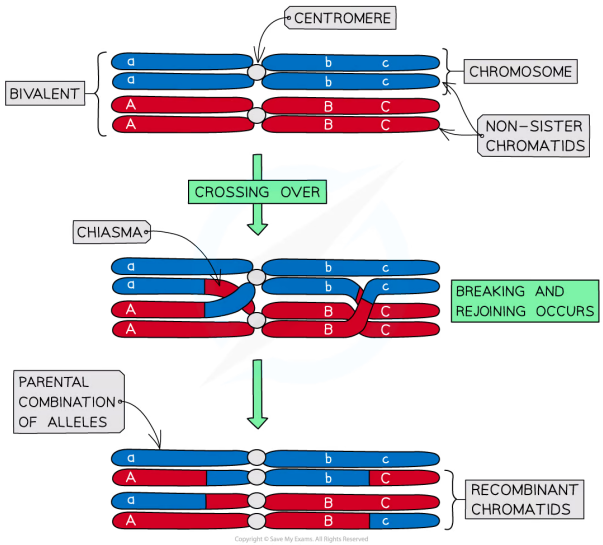
YOUR NOTES

- As Morgan continued his experiments he noticed a number of different traits in fruit flies that did not conform to Mendelian ratios as **several phenotypes occurred in much lower frequencies** than expected
- Based on this data, Morgan made two key proposals:
  - The alleles for these traits were located on **the same chromosom**e (gene linkage) meaning they did **not independently assort**
  - Linked alleles could be **unlinked via recombination** (crossing over) to produce **recombinant** offspring (offspring that have a different combination of characteristics to their parents)
    - This is due to the **crossing over** that occurs during meiosis
    - The crossing over and exchanging of genetic material **breaks the linkage** between the genes and recombines the characteristics of the parents
- Morgan also observed that the number of recombinants that resulted from crossing linked genes **varied** depending on the combination of traits
- He proposed the idea that the number of recombinants (**crossover frequency)** may be related to the **distance between two genes** on the same chromosome
  - Genes that were further apart had a **higher crossover frequency,** whereas genes closer together exhibited a lower crossover frequency
  - Morgan used this concept to create the first **gene linkage maps**
    - These maps displayed the **relative positions of genes** on chromosomes

YOUR NOTES

Identifying Recombinants in Crosses

- Genetic diagrams involving autosomal linkage often predict solely **parental type** offspring (offspring that have the same combination of characteristics as their parents)
- However in reality **recombinant** offspring (offspring that have a different combination of characteristics to their parents) are often produced
  - This is due to the **crossing over** that occurs during meiosis
  - The crossing over and exchanging of genetic material **breaks the linkage** between the genes and recombines the characteristics of the parents



The process of crossing-over results in recombinant phenotypes that can differ from the parental phenotype.

- The frequency of recombinants within a population will nearly always be less than that of non-recombinants
  - Crossing over is **random** and chiasmata form at different locations with each meiotic division
- **Recombination frequency** between two linked genes is **greater when genes are further apart** on the same chromosome
  - There are more possible locations for a chiasma to form between the genes

Identifying recombinants using test crosses

- **Test crosses** are often used to determine unknown genotypes
- Similarly, they can be used to **identify recombinant phenotypes in offspring**
- An individual is crossed with a **homozygous recessive individual (for both traits)**
  - If **any** of the offspring possess a **non-parental phenotype** then they are labelled as **recombinants**
    - These individuals have **new allele combinations** due to the process of crossing over during meiosis leading to the exchange of genetic material between chromosomes

Drawing a Punnett square to show dihybrid inheritance of linked genes

YOUR NOTES

- A number of sweet pea plants were generated by crossing double-homozygous dominant plants (***PL***)(***PL***) with double-homozygous recessive plants (***pl***)(***pl***) to produce a 100% heterozygous F_1 generation (***PL***)(***pl***) as expected
- Members of this generation were then interbred to produce the F_2 generation
- Alleles:
  - ***P*** = purple flowers, dominant to ***p*** = red flowers
  - ***L*** = long seeds, dominant to ***l*** = round seeds

Possible Gametes Table

Grandparent of F2 generation – genotypes	$\frac{PL}{PL}$ x $\frac{pl}{pl}$
Gametes	PL pl
F1 generation genotype	$\frac{PL}{pl}$
Due to crossing over, the F1 generation will produce some recombinant gametes as well as a majority of parental ones	
F1 x F2 cross – genotypes	$\frac{PL}{pl}$ x $\frac{PL}{pl}$
Gametes – parental These will be abundant	PL pl PL pl
Gametes – recombinant These will be scarce	pL Pl pL Pl

F_2 Punnet Square Showing Possible Genotypes

		Gametes from F_1 generation, parent 1			
		PL	pl	pL	Pl
Gametes from F_1 generation, parent 2	PL	$\frac{PL}{PL}$	$\frac{PL}{pl}$	$\frac{PL}{pL}$	$\frac{PL}{Pl}$
	pl	$\frac{pl}{PL}$	$\frac{pl}{pl}$	$\frac{pl}{pL}$	$\frac{pl}{Pl}$
	pL	$\frac{pL}{PL}$	$\frac{pL}{pl}$	$\frac{pL}{pL}$	$\frac{pL}{Pl}$
	Pl	$\frac{Pl}{PL}$	$\frac{Pl}{pl}$	$\frac{Pl}{pL}$	$\frac{Pl}{Pl}$

YOUR NOTES

- According to Mendelian ratios and the Punnett square, the F_2 generation should follow the typical 9:3:3:1 ratio
- However, in reality, the **frequency of recombinant gametes will be much lower** than that of parental gametes
  - This affects the resulting offspring phenotypes, with **fewer recombinant phenotypes occurring** than expected

Expected vs Predicted Phenotypes Table

EXPECTED phenotype % in F_2 offspring (9 : 3 : 3 : 1 ratio)	ACTUAL phenotype % in F_2 offspring	Phenotype	Observations
56%	70%	Purple flower, long pollen grains	More than expected
19%	5%	Purple flower, round pollen grains	Less than expected
19%	6%	Red flower, long pollen grains	Less than expected
6%	19%	Red flower, round pollen grains	More than expected

Observations

- More of the F_2 offspring than expected showed the **parental phenotypes**
- **Fewer** plants with **recombinant phenotypes** were produced than the 9:3:3:1 ratio would suggest
- The actual ratios found were referred to as '**non-Mendelian**' as they didn't follow Mendel's pattern
- However, this was not zero; **some recombinants** were still being produced

Possible Theories to Explain These Findings

- At the time, it was known that **many genes were carried on a few chromosomes**
- The idea that certain genes **share the same chromosome** was being developed by many scientists
- This suggested that genes **could be inherited together**, not by the law of independent assortment as put forward by Mendel
- The idea of linkage of genes was developed to explain the non-Mendelian ratios
  - The frequency of recombinant phenotypes is **lower** because **crossing over is a random process** and the chiasmata do not always form in the same place for each meiotic division
  - The frequency of recombinant gametes also depends on the **closeness of linkage** between the two genes
    - Genes located **close together** on a chromosome are **less likely** to be separated by crossing over
    - So recombinants of those two genes will be less frequent
- Thomas Hunt Morgan later provided proof of linkage to explain non-Mendelian ratios in his experimentation with fruit flies (Drosophila melanogaster)

Exam Tip

Remember to distinguish between sex linkage and autosomal linkage. The explanation of non-Mendelian ratios falls into the domain of autosomal linkage for IB.

YOUR NOTES

YOUR NOTES

Chi-squared Test and Dihybrid Crosses

Use of a chi-squared test on data from dihybrid crosses

- The difference between **expected** and **observed** results in experiments can be statistically significant or insignificant (happened by chance)
- If the difference between results is statistically significant it can suggest that something else is happening in the experiment that isn't being accounted for
  - For example, linkage between genes
- A statistical test called the chi-squared test determines whether there is a **significant difference** between the observed and expected results in an experiment
- The chi-squared test is completed when the data is **categorical** (data that can be grouped)

Calculating chi-squared values

- Obtain the expected and observed results for the experiment
- Calculate the difference between each set of results
- Square each difference (as it is irrelevant whether the difference is positive or negative)
- Divide each squared difference by the expected value and get a sum of these answers to obtain the chi-squared value

THE CHI-SQUARED VALUE, χ^2 IS GIVEN BY THE FORMULA

$$\chi^2 = \Sigma \frac{(O - E)^2}{E}$$

Σ = SUM OF

O = OBSERVED VALUE

E = EXPECTED VALUE

Analysing chi-squared values

- To work out what the chi-squared value means, **a table that relates chi-squared values to probabilities** is used
- If the chi-squared value represents a **larger probability than the critical probability** then it can be stated that the differences between the expected and observed results are **due to chance**
- If it represents a **smaller probability than the critical probability** then the differences in results are **significant** and something else may be causing the differences
- To determine the critical probability biologists generally use a probability of **0.05** (they allow that chance will cause five out of every 100 experiments to be different)
- The number of comparisons made must also be taken into account when determining the critical probability. This is known as the **degrees of freedom**

YOUR NOTES

Worked Example

An experiment was carried out investigating the inheritance of two genes in rabbits

- One for coat colour and one for ear length
- A dihybrid cross revealed the expected ratio of phenotypes to be 9 : 3 : 3 : 1
- Several rabbits with the double-heterozygous genotype were bred together and the phenotypes of all the offspring were recorded
- The ratio of the offspring was **not exactly what was predicted**, but was reasonably close
- In order to determine whether this was due to chance or for some other reason, the chi-squared test was used

Chi-squared Worked Example Table 1

Phenotypes & (genotypes) of F_2 offspring		Observed Number (O)	Expected Ratio	Expected Number (E)	O – E	$(O - E)^2$	$(O - E)^2 / E$
Brown coat (BB / Bb)	Long ears (EE / Ee)	87	9 : 3 : 3 : 1	90	−3	9	0.1000
Brown coat (BB / Bb)	Short ears (ee)	31		30	1	1	0.0333
Black coat (bb)	Long ears (EE / Ee)	27		30	−3	9	0.3000
Black coat (bb)	Short ears (ee)	15		10	5	25	2.5000
		160		160		$\Sigma =$	2.9333
						$\chi^2 =$	2.93

- The expected number of each phenotype is the fraction of the total number of rabbits governed by the 9:3:3:1 ratio
- These are $^9/_{16}$, $^3/_{16}$, $^3/_{16}$ and $^1/_{16}$ of 160, respectively
- In order to understand what this chi-squared value of 2.93 says about the data, a table relating chi-squared values to probability is needed

Relating Chi-Squared Values to Probability Table

YOUR NOTES

Degrees of freedom	Probability that the difference between observed and expected results is due to chance			
	0.1	0.05	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.60	5.99	9.21	13.82
3	6.25	7.82	11.34	16.27
4	7.78	9.49	13.28	18.46

- The chi-squared table displays the probabilities that the differences between expected and observed are **due to chance**
- The **degrees of freedom** can be worked out from the results. It is calculated by **subtracting one from the number of classes**
- In this example, there are four phenotypes which means four classes, 4 – 1 = 3
- This means that the values in the **third row** are important for comparison
- For this experiment, there is a **critical probability of 0.05**
- This means that **7.82** is the value used for comparison
- The chi-squared value from the results (2.93) is **much smaller than 7.82**
- 2.93 would be located somewhere to the left-hand side of the table, representing a probability much greater than 0.1
- This means that there is **no significant difference** between the expected and observed results, any differences that do occur are **due to chance**

YOUR NOTES

Worked Example

An experiment was carried out on *Drosophila* genes

- One gene for body colour ***B*** = brown, ***b*** = black
- One gene for wing shape ***W*** = straight, ***w*** = curved
- A dihybrid cross revealed the expected ratio of phenotypes to be 9 : 3 : 3 : 1
- Several *Drosophila* with the double-heterozygous genotype were bred together and the phenotypes of all the F_2 offspring were recorded
- The ratio of the offspring was **far from what was predicted**
- In order to determine whether this was due to chance or for some other reason, the **chi-squared test** was used

Chi-squared Worked Example 2 Table

Phenotypes & (genotypes) of F_2 offspring		Observed Number (O)	Expected Ratio	Expected Number (E)	O – E	$(O - E)^2$	$(O - E)^2 / E$
Brown body (BB / Bb)	Straight wing (WW / Ww)	450	9 : 3 : 3 : 1	336	114	12,996	39
Brown body (BB / Bb)	Curved wing (ww)	31		112	−81	6,561	59
Black body (bb)	Straight wing (WW / Ww)	27		112	−85	7,225	65
Black body (bb)	Curved wing (ww)	89		37	52	2,704	73
		597		597		$\Sigma =$	235
						$\chi^2 =$	235

- The expected number of each phenotype is the fraction of the total number of flies governed by the 9:3:3:1 ratio
- These are $^9/_{16}$, $^3/_{16}$, $^3/_{16}$ and $^1/_{16}$ of 597, respectively
- In order to understand what this chi-squared value of 235 says about the data, a table relating chi-squared values to probability is needed

Relating Chi-Squared Values to Probability Table

YOUR NOTES

Degrees of freedom	Probability that the difference between observed and expected results is due to chance			
	0.1	0.05	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.60	5.99	9.21	13.82
3	6.25	7.82	11.34	16.27
4	7.78	9.49	13.28	18.46

- The chi-squared table displays the probabilities that the differences between expected and observed are **due to chance**
- The **degrees of freedom** can be worked out from the results. It is calculated by **subtracting one from the number of classes**
- In this example, there are also four phenotypes which means four classes, 4 – 1 = 3
- This means that the values in the **third row** are important for comparison
- For this experiment, there is a **critical probability of 0.05**
- This means that **7.82** is the value used for comparison
- The chi-squared value from the results (235) is **much greater than** 7.82
- 235 would be located somewhere off the far right of the table, representing a very **small probability**, much less than 0.001
- This means that there is a **significant difference** between the expected and observed results, any differences that do occur are **due to a factor other than chance**
- In this case, that factor is (autosomal) **gene linkage**

Conclusion

- The alleles for black/brown body colour and straight/curved wings are linked, ie. carried on the same chromosome (autosome)

Exam Tip

When calculating a chi-squared value it is very helpful to create a table like the ones seen in the worked examples. This will help you with your calculations and make sure you don't get muddled up! You should also be prepared to suggest reasons why results might be significantly different. For example, there could be linkage between the genes being analysed.

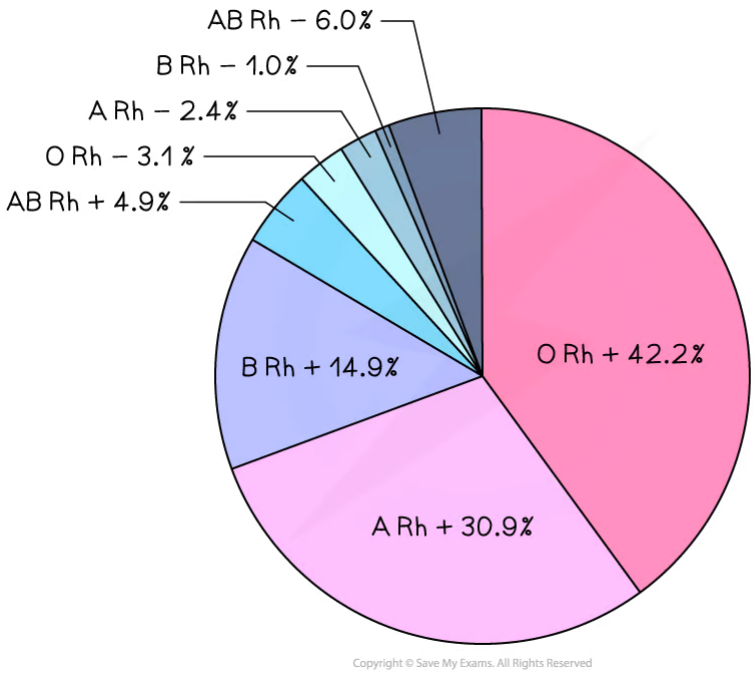
YOUR NOTES

Types of Variation

- The ways in which organisms differ from one another is called **variation**
- Variation occurs **between species**
  - In fact, species are classified based on differences between their respective members
  - This is called **interspecific variation**
- Variation can occur **within the same species**
  - Between different individuals or groups of individuals
  - This is called **intraspecific variation**
  - This suggests that only one gene is involved in governing discrete variation
  - This is called monogenic inheritance

Variation can be discrete or continuous

- Discrete variation is an example of intraspecific variation
  - Individuals fall into two or more clear-cut categories with no overlap or in-between categories
  - Blood group is an example of discrete variation
    - All human blood is either group O, A, B or AB, each with a Rhesus factor (+ or -)
- This gives just 8 distinct blood groups

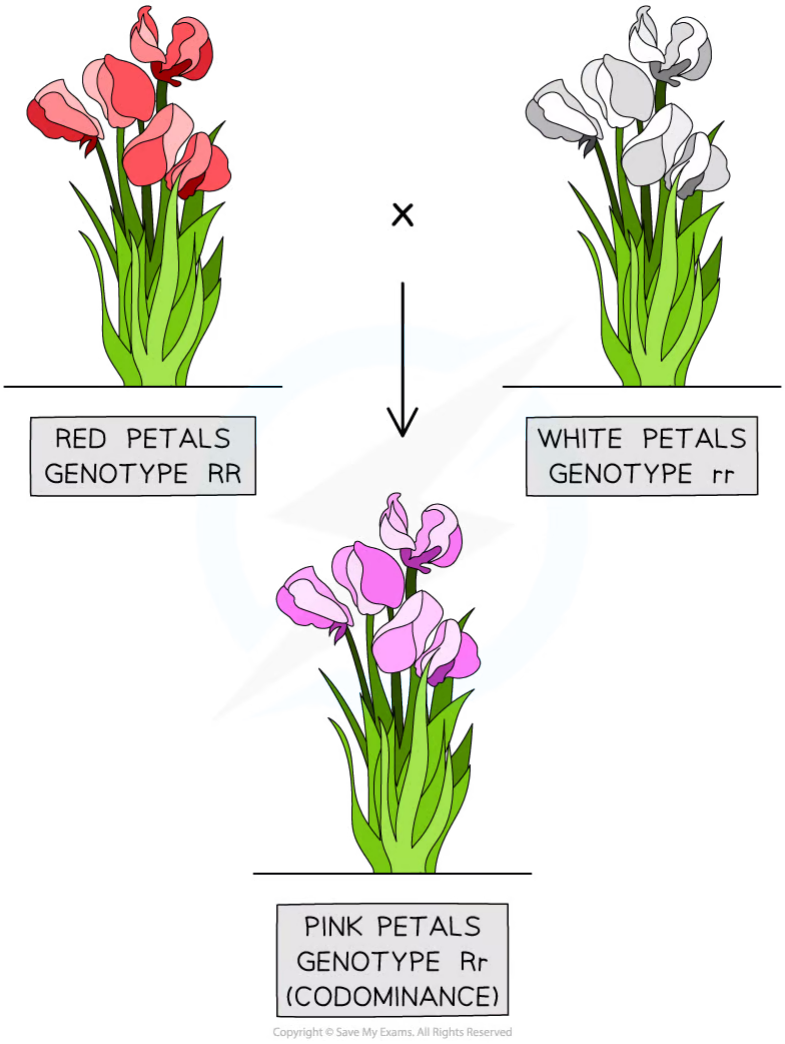


Worldwide A, B, O blood group distribution by percentage, 2019

(data varies regionally with ethnicity)

- The petal colour of snapdragons is a discrete variable; either red, white or pink with no in-between colours
- Discrete variation is sometimes referred to as **discontinuous** variation, in contrast to continuous variation

YOUR NOTES



Snapdragons display 3 main petal colours: red, white and pink, determined by a single pair of codominant alleles

This is an example of discrete variation that is solely due to genetic factors

Causes of discrete variation

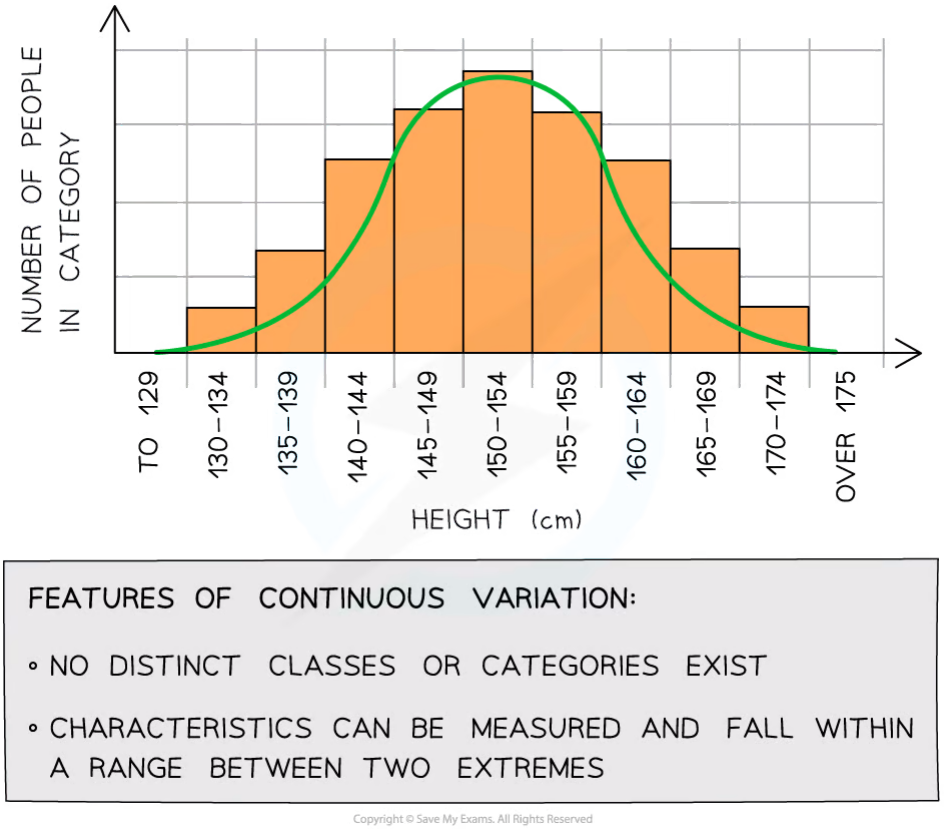
- This type of variation occurs solely due to **genetic factors**
- The environment has no direct effect
  - Phenotype = **genotype**
- At the genetic level:
  - Different **genes** have **different effects** on the phenotype
  - Different **alleles** at a single gene locus have a **large effect** on the phenotype
  - Remember diploid organisms will inherit two alleles of each gene, these alleles can be the same or different
- A good example of this is 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

Continuous Variation

YOUR NOTES

- **Continuous variation** occurs when **two or more genes** affect the final characteristic
- For example, height in humans is determined by **many genetic factors**:
  - Bone length
  - Skeletal muscle structure
  - Ability to absorb food substances effectively
  - Hormone production
  - ...As well as environmental factors like diet, exercise, prenatal nutrition, lifestyle etc
- Most characteristics are determined by more than one gene - a **polygenic** characteristic
- Even **grouped data** like shoe size appears to be discrete but in fact, peoples' feet vary continuously in size
  - Shoe size is merely a practicality for shoe manufacturers, who cannot make exactly the right-sized shoes for everybody
- Continuous variation in birth mass results in the population displaying a **normal distribution** (bell-shaped curve)
  - Of course, environmental factors can affect birth mass, eg. mother's diet, presence of a twin, smoking etc
- Continuous variation occurs when there are **quantitative differences** in the phenotypes of individuals within a population for particular characteristics
- Quantitative differences do not fall into discrete categories like in discontinuous variation
-
  - For example, the mass or height of a human is an example of continuous variation
  - Instead for these features, a **range of values** exist between two extremes within which the phenotype will fall
- 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

YOUR NOTES



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

Genetic basis of continuous variation

- This type of variation is caused by an **interaction between genetics and the environment**
- Phenotype = **genotype + environment**
- 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**

Comparison of Continuous and Discontinuous Variation Table

YOUR NOTES

Feature	Continuous variation	Discontinuous variation
Definition	Features can be measured across a complete range (from one extreme to another). Data collected are quantitative data	Features form distinct classes or categories. Data collected are qualitative data (i.e. discrete or categorical data)
Gene locus	Many loci (that may be on different chromosomes)	Usually only one but may be a very small number
Number of alleles	Many pairs of alleles as many genes contribute to the inheritance (polygenic)	Usually only one pair of alleles (monogenic) but may be a very small number
Effect on phenotype	Many intermediate phenotypes between the extremes (e.g. between shortest and tallest)	Feature either present or absent (the differences are discrete categories)
Environmental influence	Environment has a significant influence	Environment has little to no influence
Examples	Height in humans, milk yield in cattle	Ability to roll tongue, human blood groups

Environmental Influence & Variation

Polygenic traits such as human height may also be influenced by environmental factors

- Many environmental factors can affect the intraspecific variation displayed by an organism, including
  - Diet
  - Lifestyle
  - Exercise
  - Exposure to sunlight eg. tanned skin
  - Availability of soil minerals in plants
  - Human intervention eg. pruning plants, neutering animals
  - Fashion, individual preference
  - Native language and dialect (based on where an individual is brought up)
- These traits and differences have been observed in **identical twins** who were unfortunate enough to have been **separated at birth**
  - Not a practice condoned in the 21st century, but was once considered a valid investigative method
- Individuals displayed **distinct phenotypic differences** based on their diet and lifestyle differences

YOUR NOTES

10.3 Gene Pools & Speciation

10.3.1 Gene Pools

Gene Pools

- A gene pool consists of all the genes and their different alleles, present in an interbreeding population
- Some populations of the same species are geographically isolated from each other
  - So **multiple gene pools** can exist for a species
- Individuals in a population tend to have **common characteristics** and resemble each other
  - Two geographically-isolated population may have **different characteristics** whilst still being the same species
  - This is one basis for **speciation**
- A consideration of all the genes (and alleles thereof) in a population is important as that will govern the **genomes of the next generation**
  - This collection of genes and alleles is commonly called the gene pool

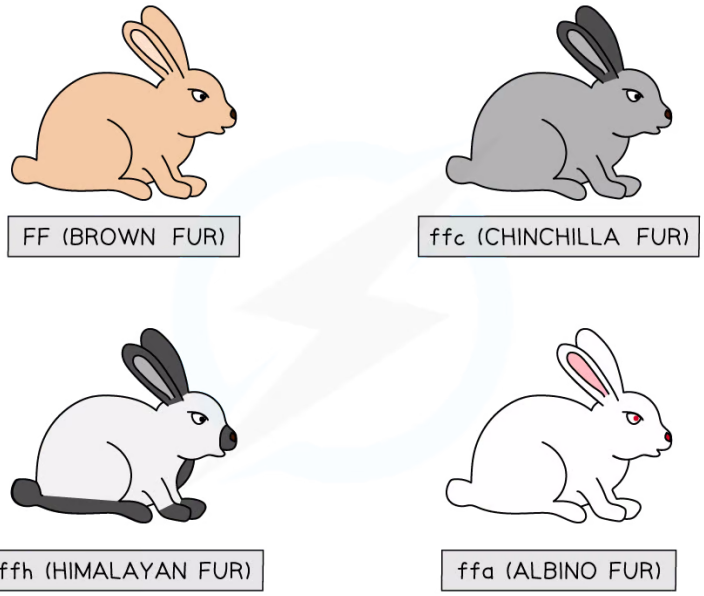
Calculating allele frequencies

- Allele frequencies are defined as the **relative abundance** of alleles for a particular gene
- Allele frequency is calculated by dividing the number of times the **allele of interest** is observed in a population by the total number of copies of **all the alleles** at that particular genetic locus in the population
  - Allele frequencies are expressed as a **number between 0.0 and 1.0** or as percentages **0 - 100%**
    - We can think of this as a **probability** that an allele chosen at random in a given gene will be a particular allele of interest
  - The frequencies of all the alleles for a particular gene **must add up to 1**

YOUR NOTES

Worked Example

In rabbits, wild-type animals have brown fur, governed by a dominant allele **F**. However, 3 recessive alleles exist, each of which gives a different fur colour and coverage.



Wild-type brown rabbits (FF) and three separate recessive-allele-containing rabbits (ffc, ffh and ffa alleles)

Four separate alleles comprise the gene pool that determines fur colour in this population:
F = brown fur
$\mathbf{f_c}$ = chinchilla fur
$\mathbf{f_h}$ = Himalayan fur
$\mathbf{f_a}$ = albino fur

The albino fur allele ($\mathbf{f_a}$) is four times more frequent in this population than the Himalayan fur allele ($\mathbf{f_h}$). Use this information to complete the table below.

Phenotype	Allele	Allele frequency
Brown	F	0.84
Chinchilla	f_c	0.06
Himalayan	f_h	
Albino	f_a	

YOUR NOTES

Step 1: Calculate the sum of the allele frequencies given

Allele frequencies ($F + f_c$) = 0.84 + 0.06 = 0.90

Step 2: Work out the sum of the remaining allele frequencies

All possible allele frequencies must add up to 1, so allele frequencies ($f_h + f_a$) = 1.0 - 0.9 = 0.1

Step 3: Apply the 4:1 ratio of albino allele: Himalayan allele given in the question

0.1 split in a 4:1 ratio is 0.08 : 0.02 for the alleles $f_a : f_h$

Step 4: Complete the table with these frequencies

Phenotype	Allele	Allele frequency
Brown	F	0.84
Chinchilla	f_c	0.06
Himalayan	f_h	0.02
Albino	f_a	0.08
		1.00

Stable gene pools

- Populations retain a **stable** gene pool under the following conditions
  - The population is **large**
  - Each individual in the population has an **equal chance of mating**
  - That matings are **random**
  - There are **no selective pressures** acting upon individuals based on their phenotype
- A stable gene pool means that a population is **not evolving**

Exam Tip

Avoid confusing **allele frequency** with **phenotype frequency**; they mean very different things. An allele can have a high frequency but if it is recessive, it may only be expressed in a minority of the phenotypes of the population.

YOUR NOTES

The Process of Evolution by Natural Selection

- Evolution requires that **allele frequencies change** with time in populations
- Evolution is defined as the **cumulative change in the heritable characteristics (genes)** of a population over time
- Organisms cannot change their species' phenotype significantly without an **underlying change in their genetic makeup**
- The factors that drive evolution are
  - **Mutations** causing new alleles to come into being
  - **Selection pressures** that favour the existence of certain alleles and oppose that of others
- A key consideration is that **evolution has no purpose**
  - There is **no conscious change** of genetic makeup in order to take advantage of changes in conditions
  - Mutations and selection pressures occur entirely at **random**
  - A species only evolves by virtue of a lucky **combination of advantageous alleles and selection pressures**
  - Organisms developing other alleles that put an affected individual at a selective disadvantage **will not survive** to reproduce and pass on those alleles
- Changes in allele frequencies can sometimes be referred to as genetic drift
- Evolution can happen within a species **before** speciation **occurs**
  - An example is the many dog breeds that all exist within the same species, *Canis familiaris*
  - Whilst many breeds have been selectively bred artificially for aesthetic reasons by humans (or to perform valuable tasks like seeing-eye dogs), most common dog breeds are capable of interbreeding to produce fertile offspring, often referred to as mongrels
- **Population size** has an effect on evolution
  - In a small population, random events such as climate change can have a **dramatic effect** on the frequency of alleles
  - By contrast, in a large population, there is **more capacity to absorb small fluctuations** in allele frequency
- When an allele is put under selective pressure, its frequency drops and sometimes falls to zero as a more advantageous allele becomes more abundant, or becomes the only allele in the gene pool

Example of changes in allele frequency driving evolution

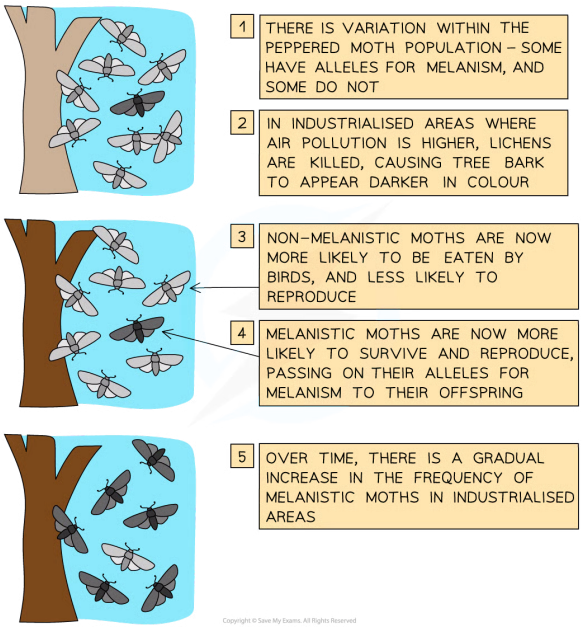
- The **peppered moth** (*Biston betularia*) is a well-documented case study of evolution by natural selection
- In the early 18th century in the northern UK, the light grey-winged form of this moth prevailed
  - These moths were **well-camouflaged** against the light-coloured bark and lichens of their host tree species
  - Black-winged moths (which carried the allele for **melanism**) fared badly as they were easy for predators (mainly birds) to spot against the light background

YOUR NOTES

- During the industrial revolution (mid-late 18th Century), soot from coal-burning factories and house chimneys coated many of the trees in the area, **turning them dark or black**
- This led to an **increase in predation of light-winged** (non-melanistic) moths whose camouflage was no longer so effective
- The dark-winged moths became the predominant variety as the **frequency of the melanism allele increased** over successive generations of moths
- Analysis of the populations throughout this period revealed the following changes in allele frequency

Allele	Dominant or Recessive	Allele Frequency / %	
		Before industrialisation (c. 1820 AD)	After industrialisation (c. 1890 AD)
 M (dark, melanism)	Dominant	0.01%	99%
 m (light, non-melanistic)	Recessive	99.99%	1%
		100%	100%

YOUR NOTES



Exam Tip

Avoid confusing **allele frequency with phenotype frequency**; these are different. In a case where the allele frequency of a recessive trait is 0.4 (40%), this means that the dominant allele must occupy 60% of the gene loci for that gene. The phenotype frequency of the recessive trait will in fact not be 40% because **two copies of the recessive allele need to combine through fertilisation** for the recessive trait to be expressed. The phenotype frequency will therefore be 0.4^2 which is 0.16 (16%). Only 16% of the population will express the recessive trait whereas 40% of the gene loci in the population contain the recessive allele.

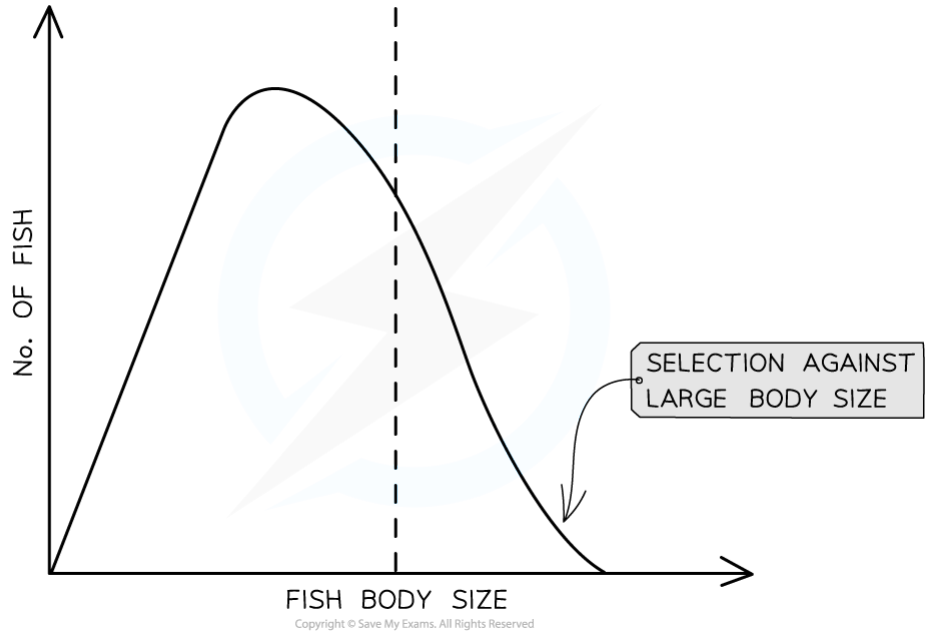
Types of Natural Selection

YOUR NOTES

- There are three main types of selection:
  - Directional
  - Stabilising
  - Disruptive

Directional selection

- The population changes **towards one extreme** of a range of variation
  - As that extreme becomes better adapted
- This tends to happen when **environmental conditions change**
- For example, a **fall in average temperatures** can affect plants that are not resistant to **frosts** (spells where the air temperature falls below 0°C)
- If there is no allele that can give the species a degree of frost resistance, then the species will **become extinct** in that habitat if cold temperatures endure for more than one generation
- If an allele exists that gives a degree of frost protection, then the species will be able to
  - Survive the frost
  - Go on to reproduce successfully
- Such an allele may code for a new protein that can lower the freezing point of water/cell contents by a few degrees and prevent the formation of damaging ice crystals
- The species has developed a **selective advantage** over other species
- A disadvantageous allele **does not have to be fatal** to an individual organism
- However, it must **prevent the individual from reproducing successfully**
  - Which is effectively the same thing from an evolutionary point of view
  - Because the allele will disappear from the gene pool as the reproductively unsuccessful individuals die
- The species can **change its genome abruptly** by directional selection

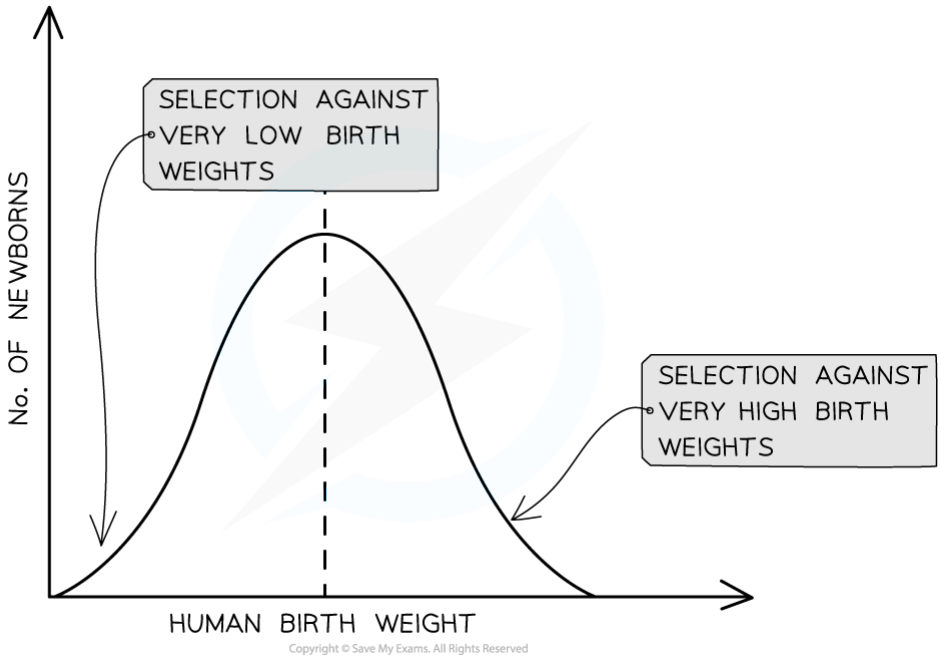


YOUR NOTES

Directional selection acting on fish body size. Increases in ocean temperatures are selecting for smaller body sizes in fish. Warmer seas cause fish metabolism to speed up and so increase their need for oxygen (oxygen levels are lower in warmer seas). Larger fish have greater metabolic needs than smaller fish, and so they feel the effect of increased temperatures more strongly.

Stabilising selection

- Selects **in favour of the average individual** in a population
- Occurs when environmental conditions are stable / do not change
- Selection tends to favour individuals with a range of alleles whose characteristic is the **most advantageous**
- Stabilising selection is the **most common form** of natural selection
- An example is the coat colour of mice
  - The colour stabilises as the one which gives the most camouflage against the surroundings eg. brown fur versus a forest floor
- Birth mass is also an example
  - Where a **normal distribution** clusters around a mean birth mass
  - Too low and too high can lead to problems of survival for an infant
- Stabilising selection
  - Discards **extreme** phenotypes
  - And instead favours the **majority of the population** that is well adapted to their local environment
  - **Decreases diversity** within a population
  - Works mostly on traits that are **polygenic**
  - Is often characterised by a **normal distribution** (a bell-shaped curve)

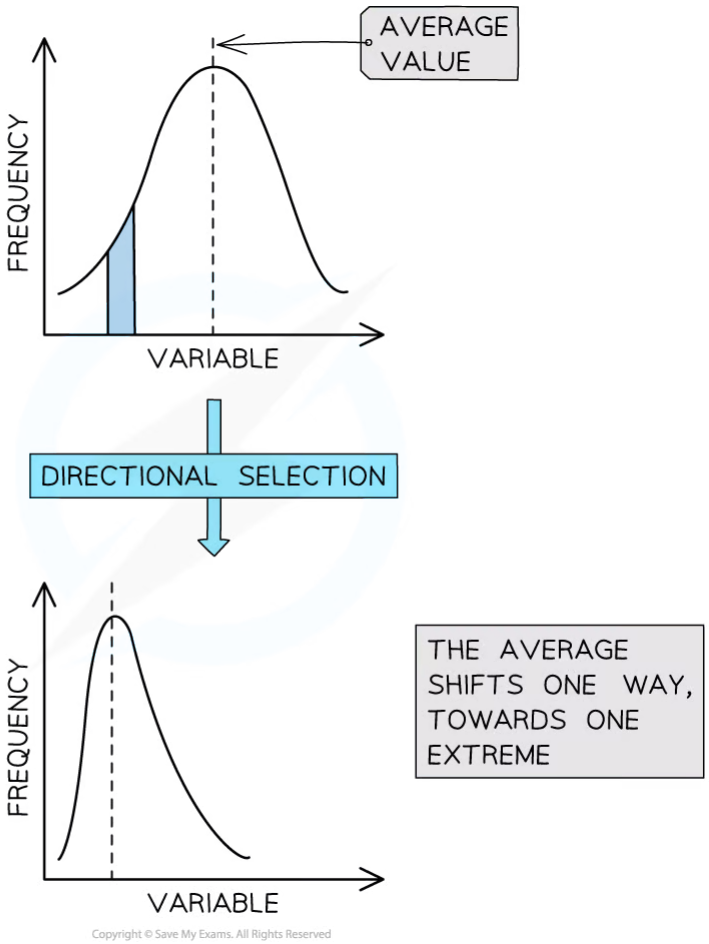


Stabilising selection on human birth weight.

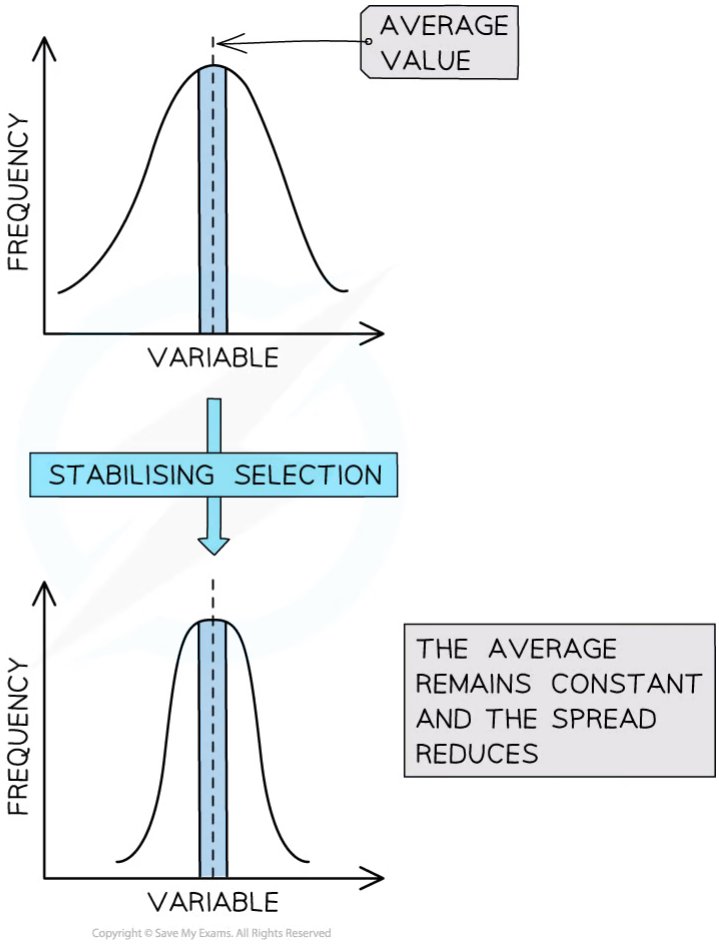
Disruptive selection

YOUR NOTES

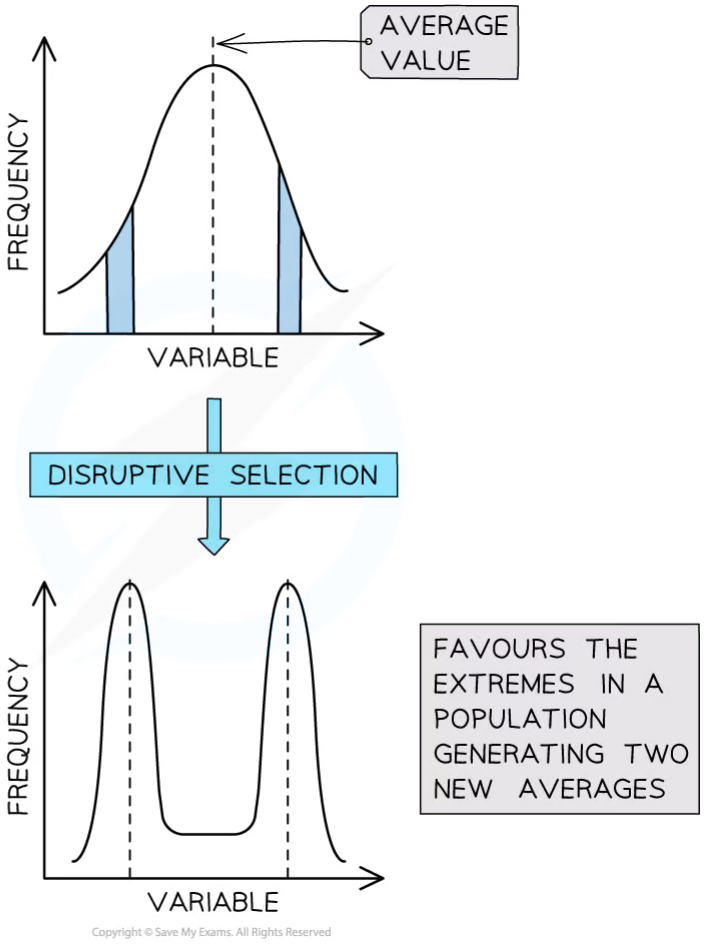
- Selects **against the average individual** in a population
- Is the rarest of the three forms of natural selection
- Like directional selection, disruptive occurs when **habitats or resources undergo a change**
- Disruptive selection can lead to the **formation of an entirely new species** (speciation)
  - For this reason, is sometimes referred to as '**diversifying** selection'
- **Darwin's finches** in the Galápagos Islands are one of the best-known examples
- Fifteen different species evolved from a common ancestor
- **Multiple types of beaks** have adapted to different food sources over time
- On one island, Santa Cruz
  - Ground finches eat more seeds and some arthropods
  - Tree finches eat more fruits and arthropods
  - Vegetarian finches feed on leaves and fruit
  - Warblers typically eat more arthropods
- When food is **abundant**, their **diets can overlap**
- When food is scarce, these specialisations give each species the ability to compete for a certain type of food better than other species
- This helps each species to occupy its own niche



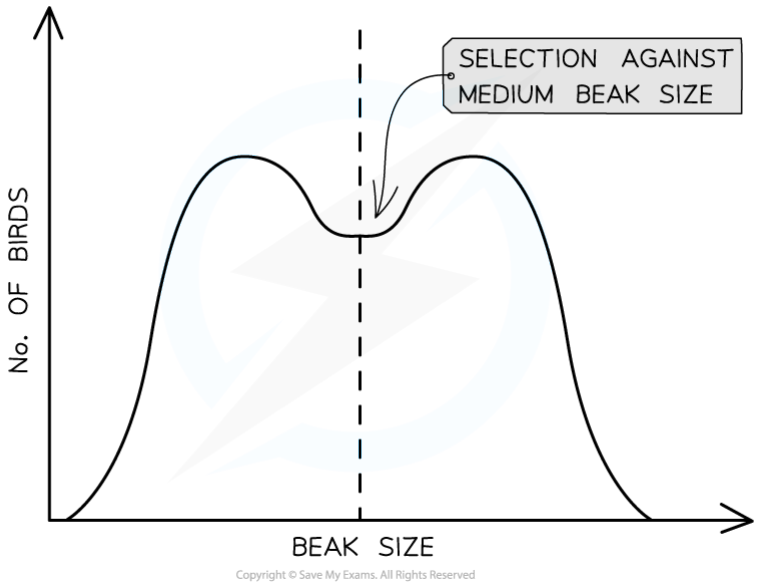
YOUR NOTES



YOUR NOTES



Summary of the 3 main forms of natural selection and their effects on the average phenotype of a population



Disruptive selection acting on beak size in a bird population

YOUR NOTES

Exam Tip

Become familiar with the shapes of the graphs above. They can help you answer questions about the type of selection that is occurring in a population.

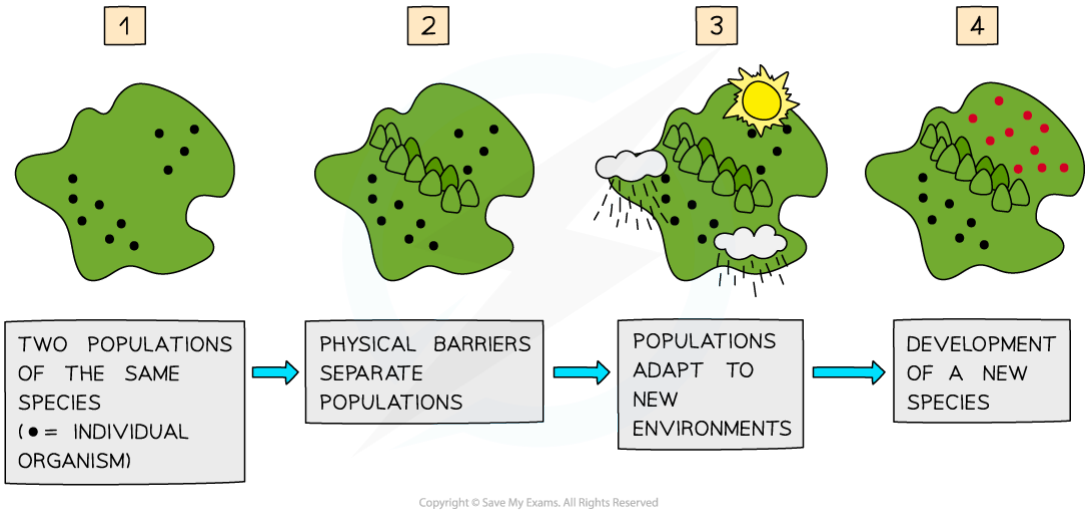
YOUR NOTES

Reproductive Isolation

- Reproductive isolation of populations can be temporal, behavioural or geographic
- A population of a species can become **isolated from its peers** by various means
  - A **barrier** forms, meaning no interbreeding occurs between the two populations
- For many generations, there may be **little or no variation of the gene pool** within that population
- The nature of the barrier between populations can be temporal, behavioural or geographic
  - Temporal (ie. seasonal)
    - Different groups have **reproductive cycles at different times of the year** so cannot interbreed
    - eg. Changes in **flowering patterns** of plants in different seasons
  - Behavioural
    - Different groups have different rituals or patterns of behaviour meaning that the groups do not recognise each other as 'self'
    - eg. Courtship rituals
  - Geographical
    - eg. When natural or man-made barriers form between two parts of a population, such as a river, freeway, or mountain range
- Over a longer period, the formation of new species (**speciation**) occurs by reproductive isolation ie. separating parts of the population into independently-breeding groups
- Natural selection will **take different paths** according to the differences of biotic and abiotic factors on either side of the divide

Sympatric vs allopatric speciation

- Temporal and behavioural speciation are both examples of **sympatric speciation**, in which separately-developed species can coexist in the **same geographical area** whilst occupying different niches
- Geographic speciation is sometimes called **allopatric speciation**, in which two separate species diverge with **complete spatial separation**



Geographical isolation can lead to speciation.

Exam Tip

A typical exam question will ask you to identify which form of natural selection is taking place from a set of data in the question. Alternatively, you may have to give reasons from the data about why a particular example of natural selection is identified as one of the three forms. Think about how the 'average' individual in a population is affected and that will lead you towards the correct answer.

YOUR NOTES

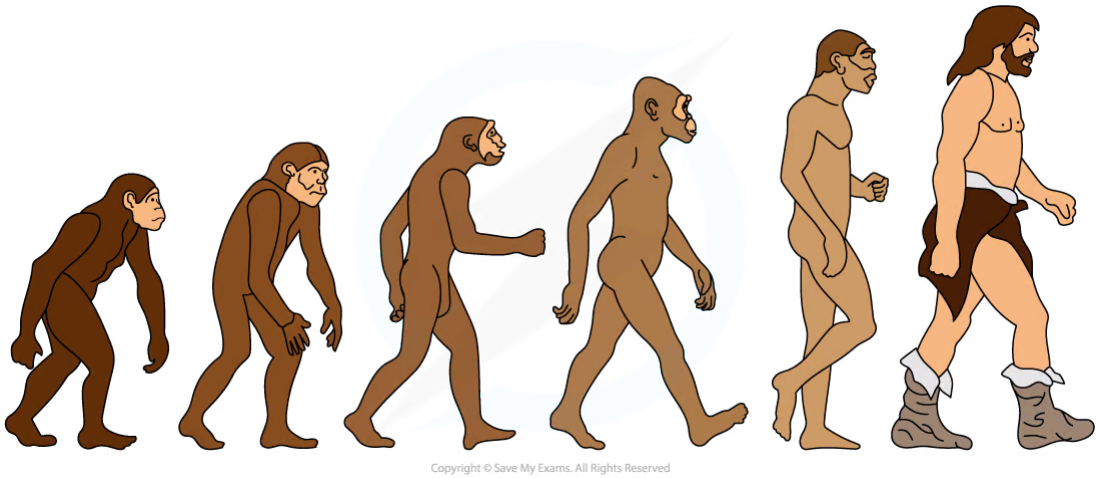
YOUR NOTES

Gradualism & Speciation

- Speciation can be defined as the emergence of new and distinct species that are reproductively isolated from other separate species
- Two theories exist to explain the emergence of new species
  - Gradualism
  - Punctuated Equilibrium

Gradualism

- Speciation due to divergence of isolated populations can be **gradual**
- Large changes between species occur due to the **culmination of many small changes** that **accumulate** over time
- Because of the long period of time in which life has existed on Earth (approx 3.5 billion years), one might expect that organisms speciated gradually from their ancestors
- Evidence exists in the fossil record to show that **patterns of evolution can follow the geological cycle**, which consists of long, slow changes that take place over thousands and millions of years
- Charles Darwin originally subscribed to the point of view of gradualism, having observed **vestigial structures** in the fossil record
  - Vestigial structures are observable characteristics that have **no apparent function**
  - They are **residual parts** from a past ancestor that are still inherited but have fallen into disuse
  - Examples of vestigial structures include the **human appendix** and the **wings of flightless birds**

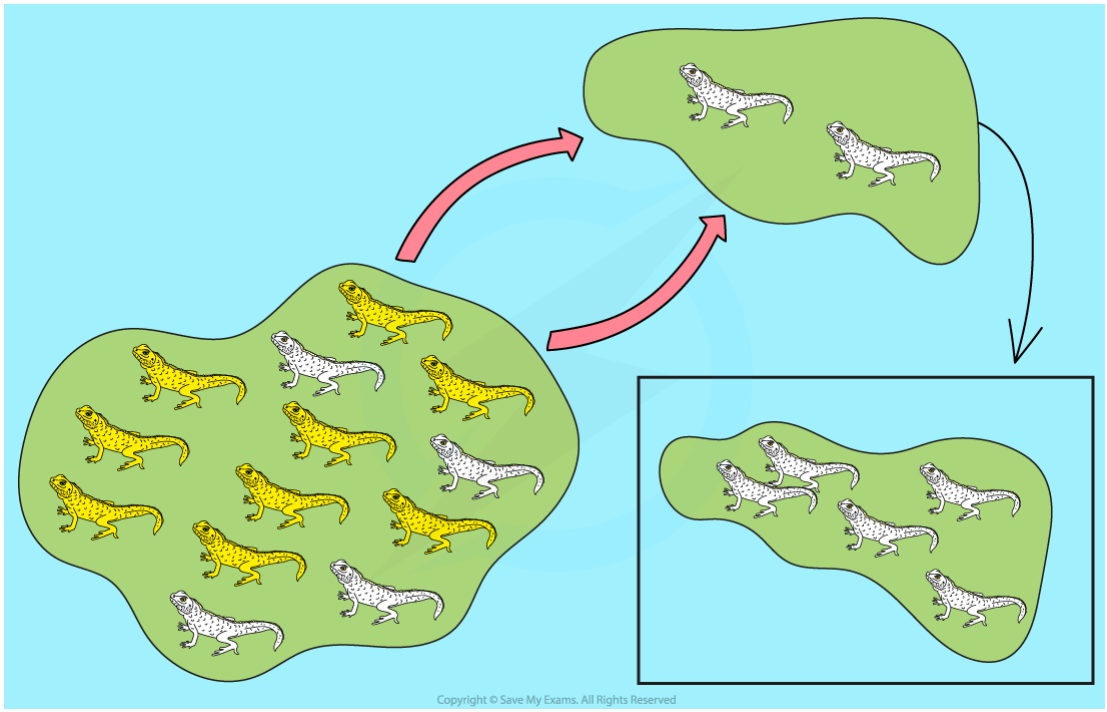


The iconic image of evolution implies gradualism as humans and chimpanzees both evolved from a common ancestor via intermediate species

Punctuated Equilibrium

YOUR NOTES

- Speciation can occur **abruptly**
- Punctuated equilibrium implies long periods without appreciable change and short periods of **rapid** evolution
- In the late 19th century, palaeontologists (scientists specialising in the study of life forms that existed in past geological periods) began to notice **anomalies in the fossil record** that **cast doubt** on Darwin and others' theories of gradualism
- One such scientist was William Bateson (who along with Reginald Punnett first observed non-Mendelian inheritance patterns)
- Breaks occurred in the fossil record that revealed **no intermediate species**
- Fossil appeared **relatively unchanged** for long periods of time yet **changed abruptly** at other times
- Sudden **mass extinctions** were observed
    - Cataclysmic events such as huge **volcanic eruptions**, **meteor strikes** and **large-scale gaseous changes to the atmosphere** can cause mass extinctions
    - Some members of the populations that are not adversely affected may **survive** the event
    - These can restart reproduction with a **reduced gene pool**
    - This is called the **Founder Effect**



The Founder Effect as shown by lizards. If the original island was destroyed and only the white (recessive phenotype) lizards move to the new island and so the whole population ends up having the white phenotype.

Exam Tip

An analogy for **punctuated equilibrium** is a pool-drop river that features long stretches of calm, slow-flowing water punctuated by rapids and waterfalls. **Gradualism** would be represented by a river that flows smoothly down a shallow, uninterrupted gradient out to the ocean.

YOUR NOTES

YOUR NOTES

Polyploidy & Speciation

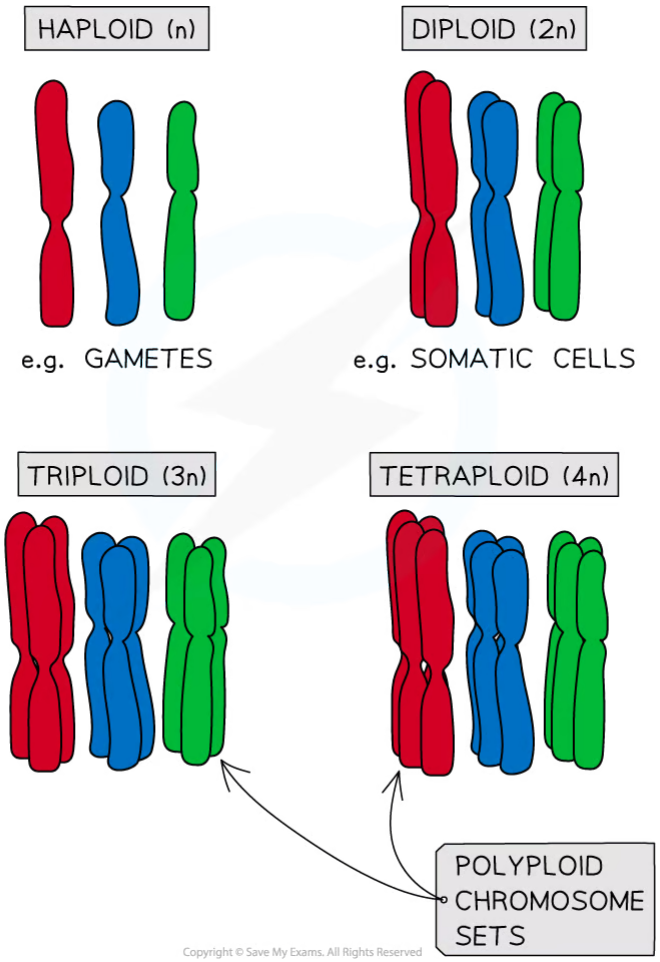
NOS: Looking for patterns, trends and discrepancies—patterns of chromosome number in some genera can be explained by speciation due to polyploidy

- When patterns and trends are observed in nature, scientists seek to find explanations that fit with these observations
- When exceptions to accepted trends are observed in the natural world, it can sometimes mean that established modes of thinking are incorrect, so it is important to consider discrepancies carefully

Polyploidy

- So far, speciation has been explored in the context of mutations and changes to an organism's **existing genome**
  - Where the number of chromosomes remains 2*n* or 2 x the haploid number, but the chromosomes' base sequences alter as new alleles form
- Speciation can also occur through **polyploidy**
- Polyploidy
  - Occurs when an organism has **more than two sets** of homologous chromosomes
  - Is more common in **plants** than in animals
  - Can result from **chromosomal mis-events** eg. abnormalities in mitosis or more commonly, in meiosis
    - For example, the fertilisation of an egg by more than one sperm, or by failure of chromosomes to separate in meiosis I
- A diploid gamete can be formed, which can fertilise with a haploid gamete and produce fertile offspring
- Polyploidy creates a separate taxonomic category within species, often called **breeds or varieties**
- Patterns in chromosome number between organisms (diploid, triploid, tetraploid etc) have been used to explain speciation
- Polyploidy can result in **sympatric speciation** as polyploid and diploid counterparts can coexist in the same geographical area

YOUR NOTES



Chromosome sets showing haploid, diploid and polyploid sets

Uses of polyploidy

Application: Speciation in the genus *Allium* by polyploidy

- Many crop species have been created to be polyploid
  - For example, species in the *Allium* genus
    - Such as onion, garlic, shallots, leeks and chives
    - *Allium porrum* is the cultivated leek and is tetraploid and fertile, so has many advantages over non-polyploid counterparts
- Wild onion (*Allium canadense*) has a diploid number of $2n = 14$ although polyploid varieties have been generated with $2n = 28$
- The common onion, (*Allium cepa*) has naturally-occurring polyploid varieties, some of which have been cultivated for agricultural use
- Polyploidy **increases allelic diversity** and **permits novel phenotypes** to be generated
  - Having **multiple copies of the same gene** reduces the risk of recessive mutations causing detrimental effects
  - Novel phenotypes can **include improved flavour** and aroma for cooking, a **greater yield** for farmers and **improved pest resistance**
- It also leads to **hybrid vigour** (the tendency of cross-bred individuals to show superior characteristics to those of their parents)

YOUR NOTES

Populations & their Allele Frequencies

Comparison of allele frequencies of geographically isolated populations

- Allele frequency is a term that assigns a **relative frequency** of an allele at a particular gene locus
  - Alleles can vary from each other by as little as one nucleotide
- When a degree of geographic **separation** exists between two populations, this can cause differences in the frequencies of alleles to emerge
- Human allele frequencies vary by **geography** and **ethnicity**
  - Examples of clear-cut allele frequency differences are rare in human populations because of the ease of **travel** and **interbreeding**
  - This leads to a scarcity of **truly isolated** populations
- If there are more than one allele in existence for a particular gene, the respective **allele frequencies must add up to 1**
- Online databases list the frequencies of human alleles
- Alleles are sometimes referred to as **polymorphisms** which just means many (poly-) different forms (-morphisms) of a gene
  - The most common type is called a **single nucleotide polymorphism** (SNP)
- Mathematical formulae such as the **Hardy-Weinberg formula** can be used to calculate phenotype frequencies from allele frequencies and *vice versa*
- Comparing allele frequencies can provide information for
  - Identifying **genetic associations** with particular diseases
  - Estimating the **number of individuals with disease susceptibility** within a population
  - Estimating the level of **drug resistance** in a population
  - Performing **evolutionary and anthropological studies** (eg. tracing the history of humans through time)

Basis of allele frequency analysis

- For any polymorphism, each individual carries **two alleles per locus**
  - One is inherited from the mother, the other from the father
  - Exception - this does not apply to alleles present on the X or Y chromosome
- Within a population, there are **twice as many total alleles** as there are individuals
- **Homozygous** individuals each contribute **two** of that allele to the total number of that particular allele
- **Heterozygous** individuals each contribute **one** of a particular allele to the total number of that allele
  - For example, if there are eight individuals with the **ZZ** genotype, they contribute 16 **Z** alleles. Thirty-four **Zz** heterozygous individuals contribute a total of 34 **Z** alleles and 34 **z** alleles to the total

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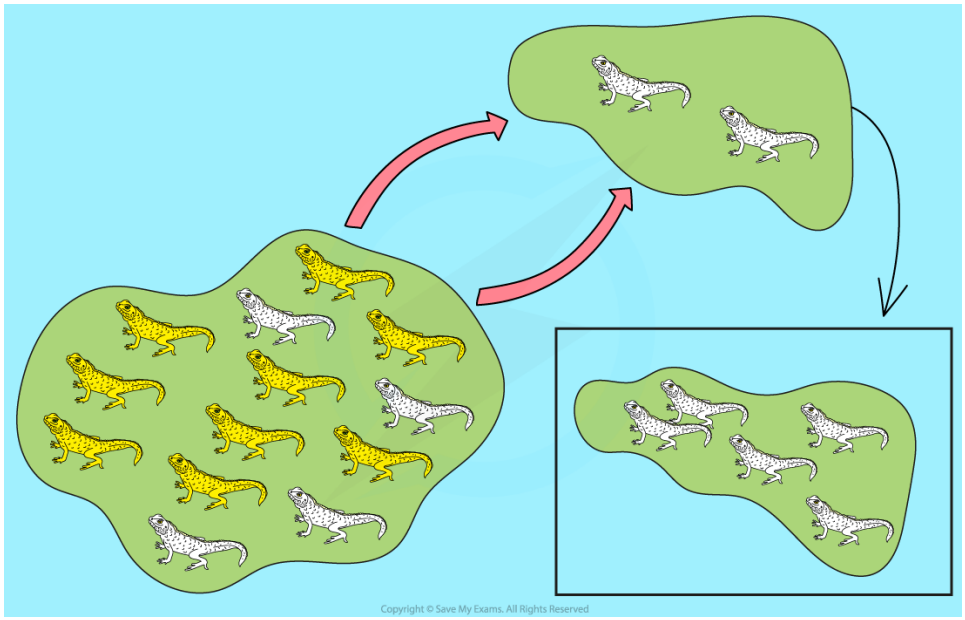
Worked Example

Consider the following isolated island populated by a certain species of lizard. On this island, the ratio of white lizards to yellow lizards is 4:9. The yellow pigmentation is caused by a dominant allele, **Y**. Lizards possessing the homozygous recessive genotype (**yy**) are white in colour.



Distribution of lizards on the island before and after the geological event.

A geological event caused the island to become divided in two; this is shown by the red arrows in the diagram above. One island sank below the ocean, killing all its inhabitants, whilst the other survived. Only two white lizards made it to the surviving half of the island. Fortunately, these were male and female and they were able to begin the recolonisation of the small island.

Calculate the allele frequencies of **Y** and **y** before and after the geological event.

Step 1: Calculate the phenotype frequencies

If 4/13 are the white lizards, the phenotype frequency of **yy** is $4 \div 13 = 0.3077$

Step 2: Calculate the allele frequency of y (from the phenotype frequency in Step 1)

The probability of two y alleles coming together through fertilisation is given by (**q** x **q**) or $\mathbf{q}^2$,
where **q** is the allele frequency of the recessive allele, **y**
Therefore, **q** is the square root of the white phenotype frequency

YOUR NOTES

$$q = \sqrt{\frac{4}{13}} = 0.5547$$

Step 3: Subtract this from 1 to find the allele frequency of the dominant allele, Y

1 - 0.5547 = 0.4453

Frequency of **Y** allele before = 0.4453 or 44.5%
Frequency of the **y** allele before = 0.5547 or 55.5%
Frequency of **Y** allele after = 0 or 0%
Frequency of the **y** allele after = 1.0 or 100%

Exam Tip

Mathematical derivations of allele frequencies are not required for your exams, although it helps to appreciate that the sum total of all the allele frequencies must add up to 1, in order to appreciate the variation within a species.