Edexcel GCSE Biology

Your notes

Inheritance

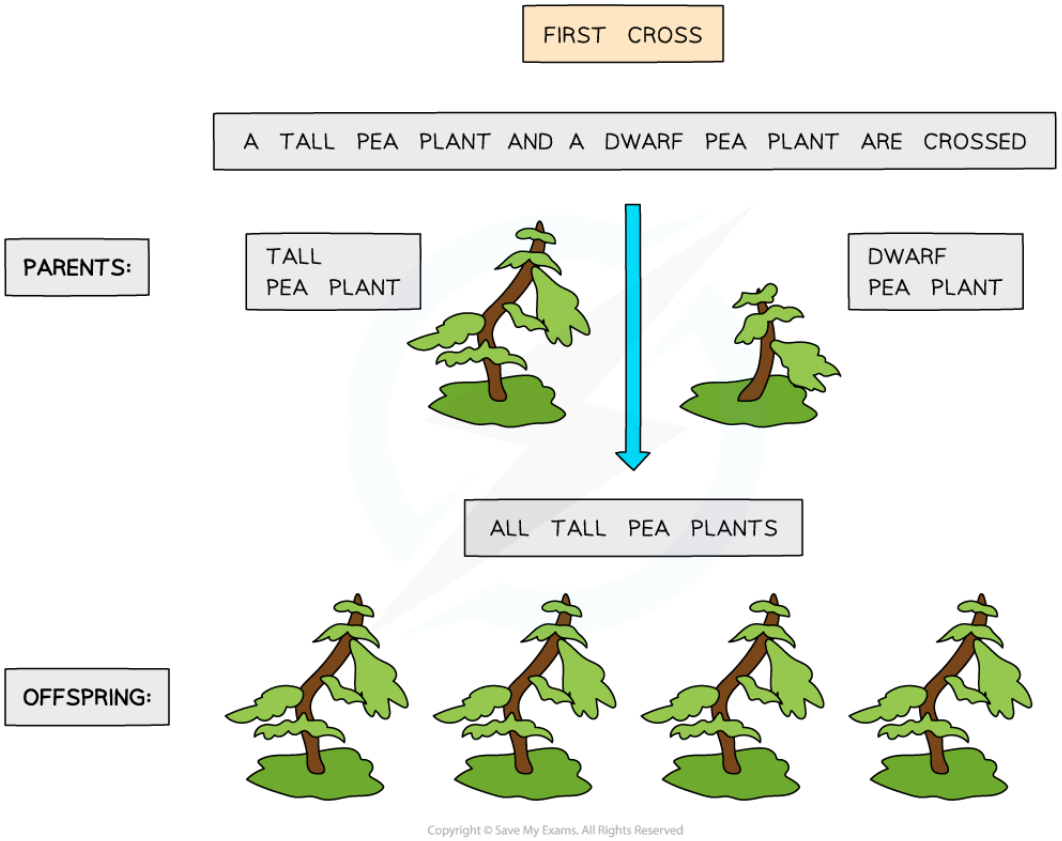
Contents

Mendel's Work

Your notes

Mendel's Work

- Gregor Mendel was an Austrian monk
- He studied mathematics and natural history at the University of Vienna
- In the mid-19th century, Mendel carried out breeding experiments on plants
- He studied how **characteristics were passed on between generations of plants**
- For example, he conducted studies with pea plants and looked at how the height characteristic was inherited
  - In the first experiment, he crossed a tall pea plant with a dwarf pea plant
  - In the second experiment, he crossed two of the tall **offspring** together



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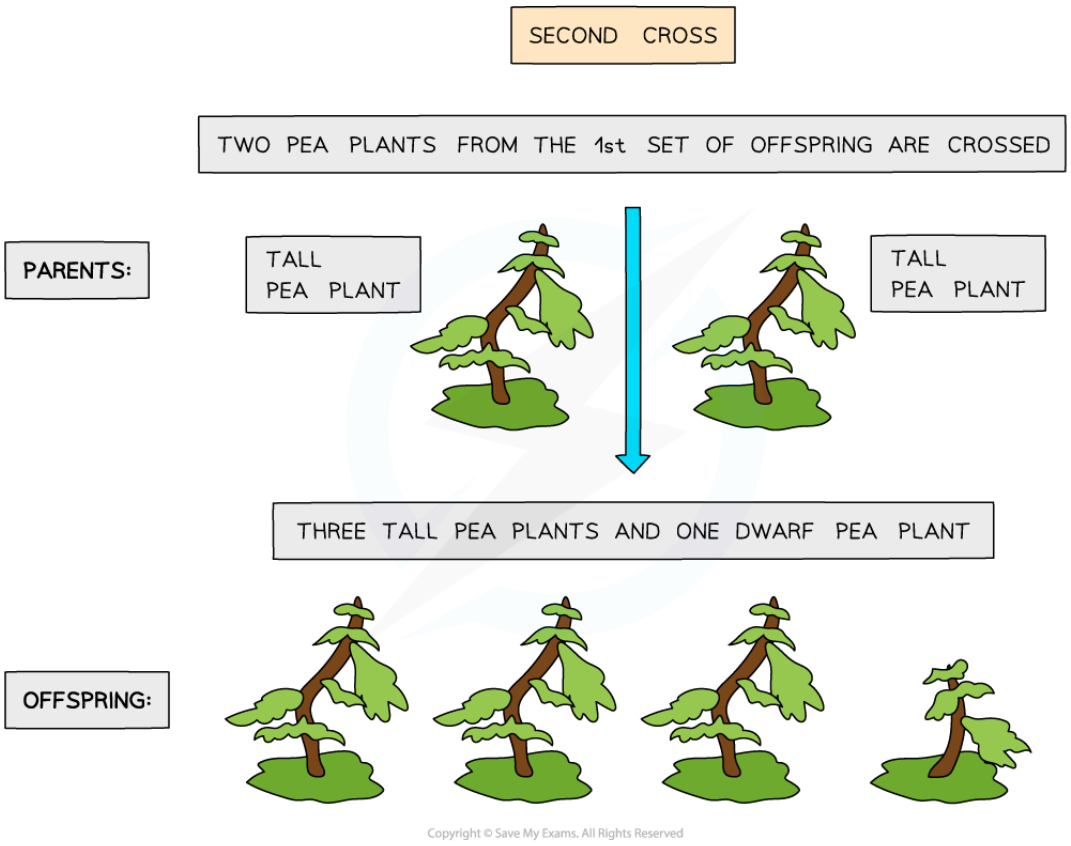


The pea plant crosses were originally carried out by Mendel to investigate the inheritance of characteristics

- One of his observations was that **the inheritance of each characteristic is determined by 'units' that are passed on to descendants unchanged**
- Using the example above, Mendel showed that height in pea plants was the result of separately inherited **'hereditary units'** passed down from each parent plant to the offspring plants – this particular experiment showed that the 'unit' for tall plants (T) was dominant over the 'unit' for short plants (t)
- He also carried out experiments to show how other characteristics of pea plants are inherited in the same way
  - E.g. Flower colour

Mendel's conclusions

- Three important conclusions about hereditary in plants were reached

- Characteristics are determined by '**hereditary units**' and these hereditary units are passed on **from parent to offspring** unchanged
- The offspring receives **one 'hereditary unit' from each parent**
- Hereditary units can be **dominant or recessive** (a dominant characteristic is always expressed when present)


Your notes

Understanding Mendel's work

- His work eventually provided the foundation for modern genetics
- The importance of Mendel's discovery was not recognised until after his death:
  - His studies were totally new to science in the 19th century
  - There was no knowledge of the mechanisms behind his findings (**DNA, genes and chromosomes had not been discovered yet**)

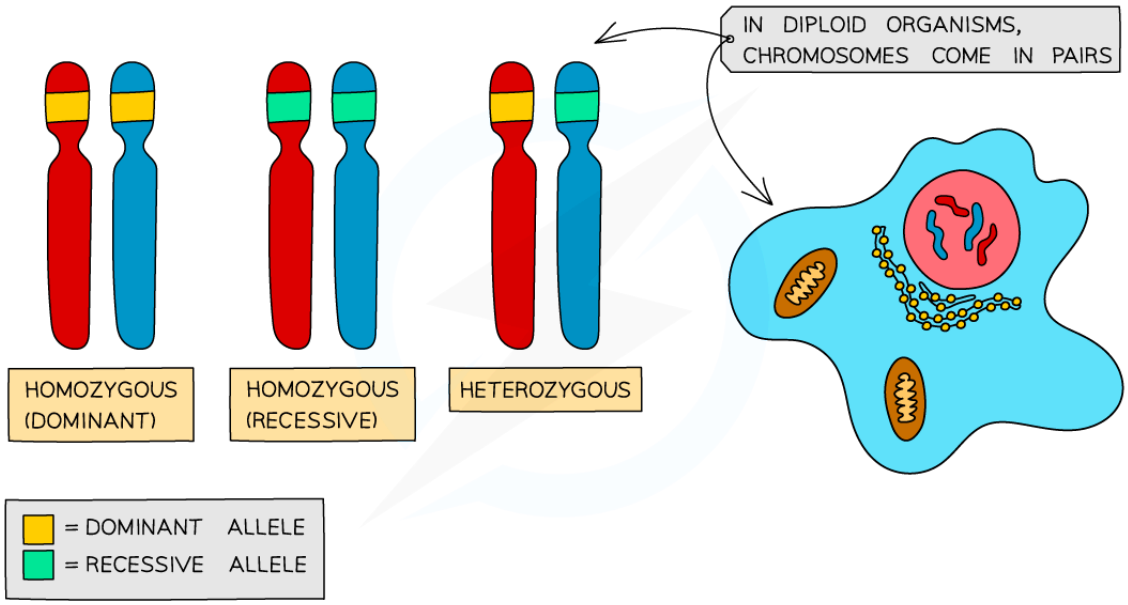
Key Definitions


Your notes

Key Definitions

- A **gene** is a short length of DNA found on a chromosome that codes for a particular **characteristic** (expressed by the formation of different proteins)
- **Alleles** are variations of the same gene
  - As we have two copies of each chromosome, we have two copies of each gene and therefore two alleles for each gene
  - One of the alleles is inherited from the mother and the other from the father
  - This means that the alleles do not have to 'say' the same thing
  - For example, an individual has two copies of the gene for eye colour but **one allele could code for brown eyes and one allele could code for blue eyes**
- The **observable characteristics** of an organism (seen just by looking - like eye colour, or found – like blood type) is called the **phenotype**
- The **combination of alleles that control each characteristic** is called the **genotype**
- Alleles can be **dominant** or **recessive**
  - A dominant allele **only needs to be inherited from one parent** in order for the characteristic to show up in the phenotype
  - A recessive allele needs to be i**nherited from both parents in order** for the characteristic to show up in the phenotype.
  - If there is only one recessive allele, it will remain hidden and the dominant characteristic will show
- If the two alleles of a gene are the same, we describe the individual as being **homozygous** (homo = same)
- An individual could be **homozygous dominant** (having two copies of the dominant allele), or **homozygous recessive** (having two copies of the recessive allele)
- If the two alleles of a gene are different, we describe the individual as being **heterozygous** (hetero = different)
- When completing genetic diagrams, alleles are abbreviated to single letters
  - The dominant allele is given a **capital letter** and the recessive allele is given the same letter, but **lower case**

Your notes



Alleles are different forms of the same gene. You can only inherit two alleles for each gene, and they can be the same (homozygous) or different (heterozygous).

- We **cannot always tell the genotype** of an individual for a particular characteristic just by looking at the phenotype – a phenotype associated with a dominant allele will be seen in both a dominant homozygous and a dominant heterozygous genotype
- If two individuals who are both **identically homozygous** for a particular characteristic are bred together, they will **produce offspring with exactly the same genotype and phenotype as the parents** - we describe them as being **'pure breeding'** as they will always produce offspring with the same characteristics
- A **heterozygous** individual can pass on **different alleles** for the same characteristic each time it is bred with any other individual and can therefore **produce offspring with a different genotype and phenotype than the parents** - as such, **heterozygous individuals are not pure breeding**

Key Terms & Definitions for Genetic Inheritance Table

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

Predicting Genetic Inheritance


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Monohybrid Inheritance

- **Monohybrid inheritance** is the inheritance of characteristics controlled by a single gene
- It can be investigated using a **genetic diagram** known as a **Punnett square**
- A Punnett square diagram shows the **possible combinations of alleles** that could be produced in the offspring
- From this, the **ratio** of these combinations can be worked out
- Remember the **dominant allele** is shown using a capital letter and the **recessive allele** is shown using the same letter but lower case

Pea plants

- Pea plants were used by the scientist Mendel to investigate monohybrid inheritance
- The height of pea plants is controlled by a single gene that has two alleles: tall and short
- The tall allele is dominant and is shown as **T**
- The small allele is recessive and is shown as **t**

A pure breeding short plant is bred with a pure breeding tall plant

- The term '**pure breeding**' indicates that the individual is homozygous for that characteristic

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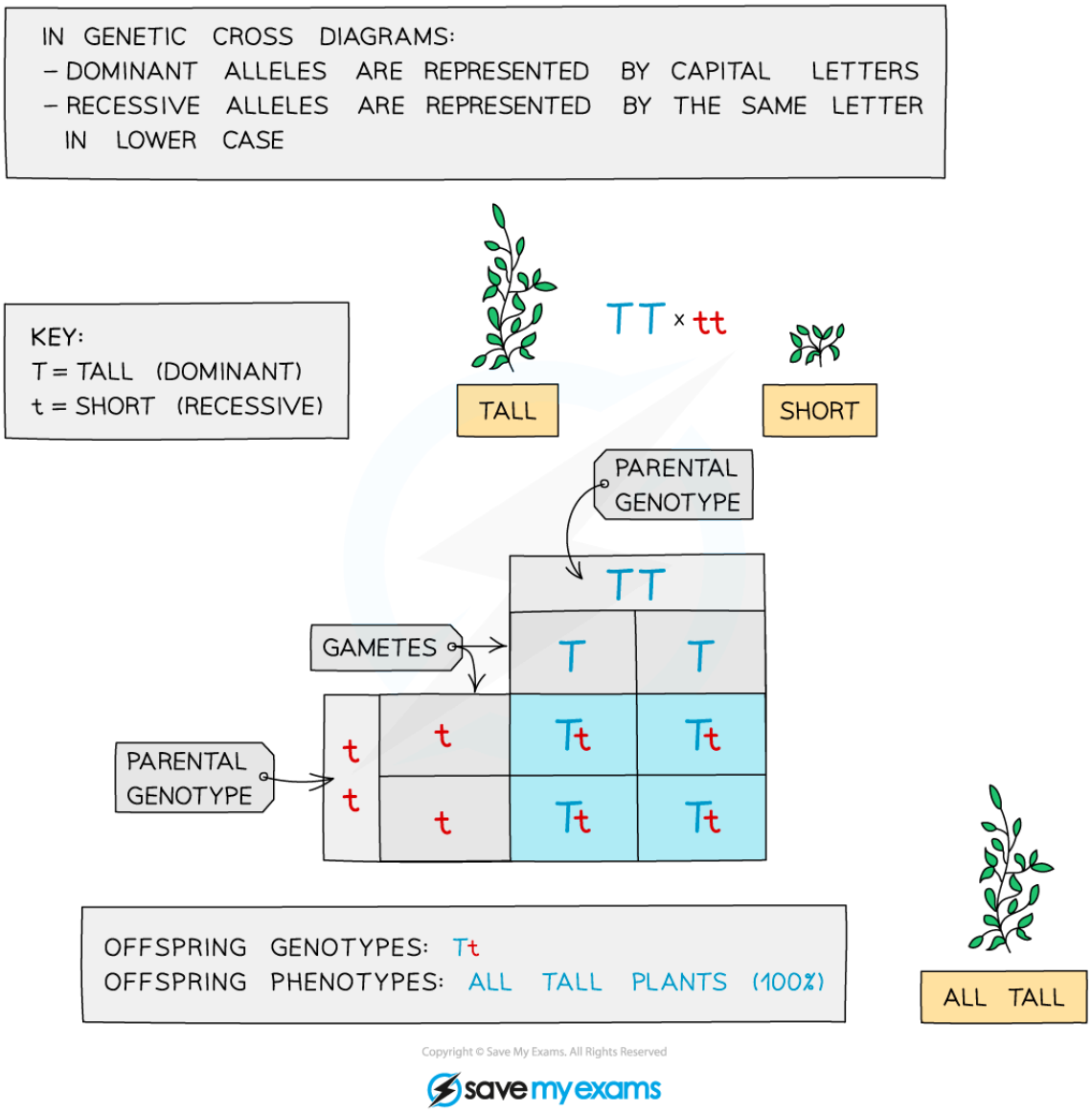

A pure-breeding genetic cross in pea plants. It shows that all offspring will have the tall phenotype.

Crossing the offspring from the first cross

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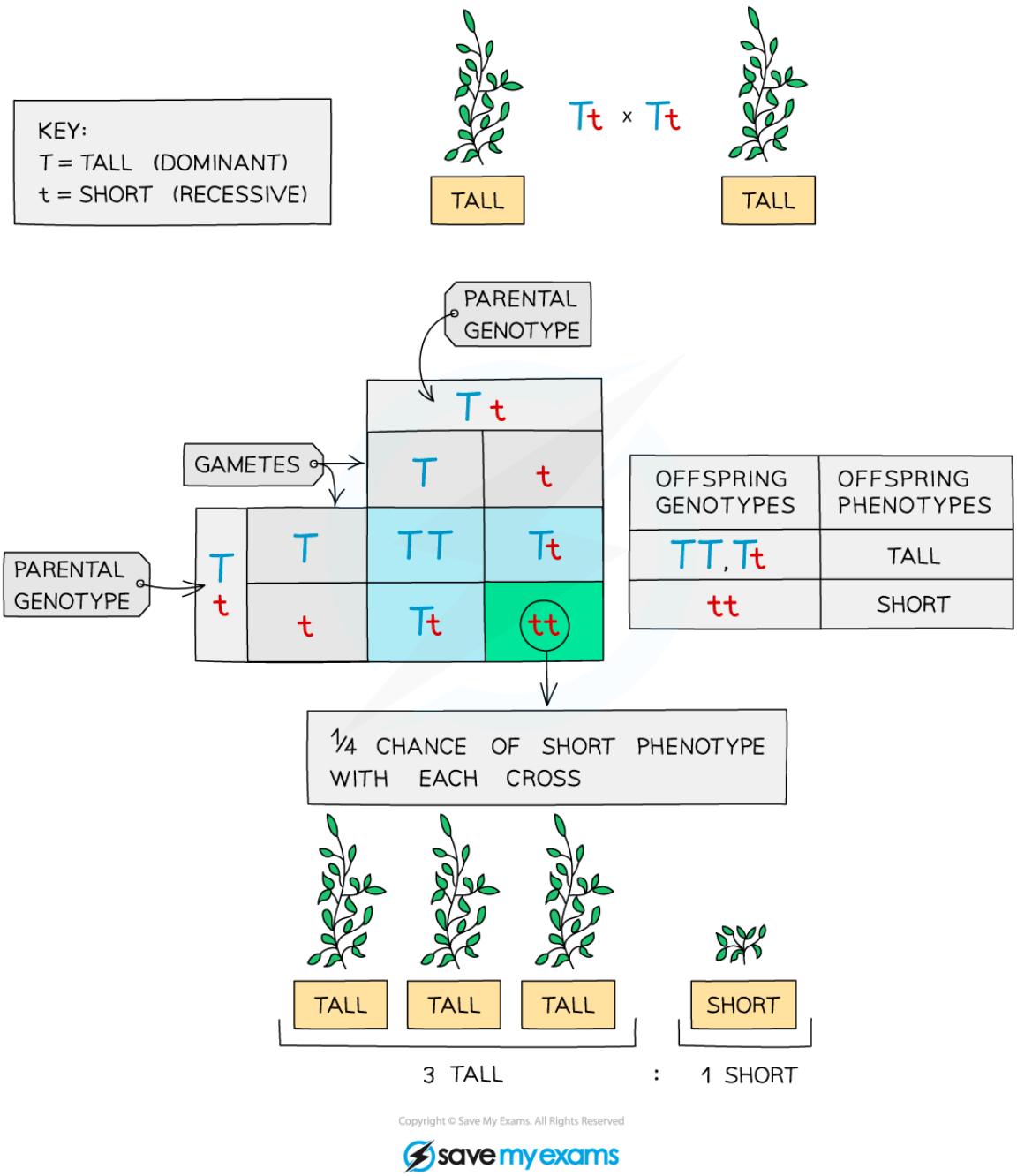

A genetic cross diagram (F2 generation). It shows a ratio of 3 tall : 1 short for any offspring.

- All of the offspring of the first cross have the same genotype, **Tt** (heterozygous), so the possible combinations of offspring bred from these are: **TT** (tall), **Tt** (tall), **tt** (short)

- There is more variation in the second cross, with a **3:1** ratio of tall : short
- The F2 generation is produced when the offspring of the F1 generation (pure-breeding parents) are allowed to interbreed

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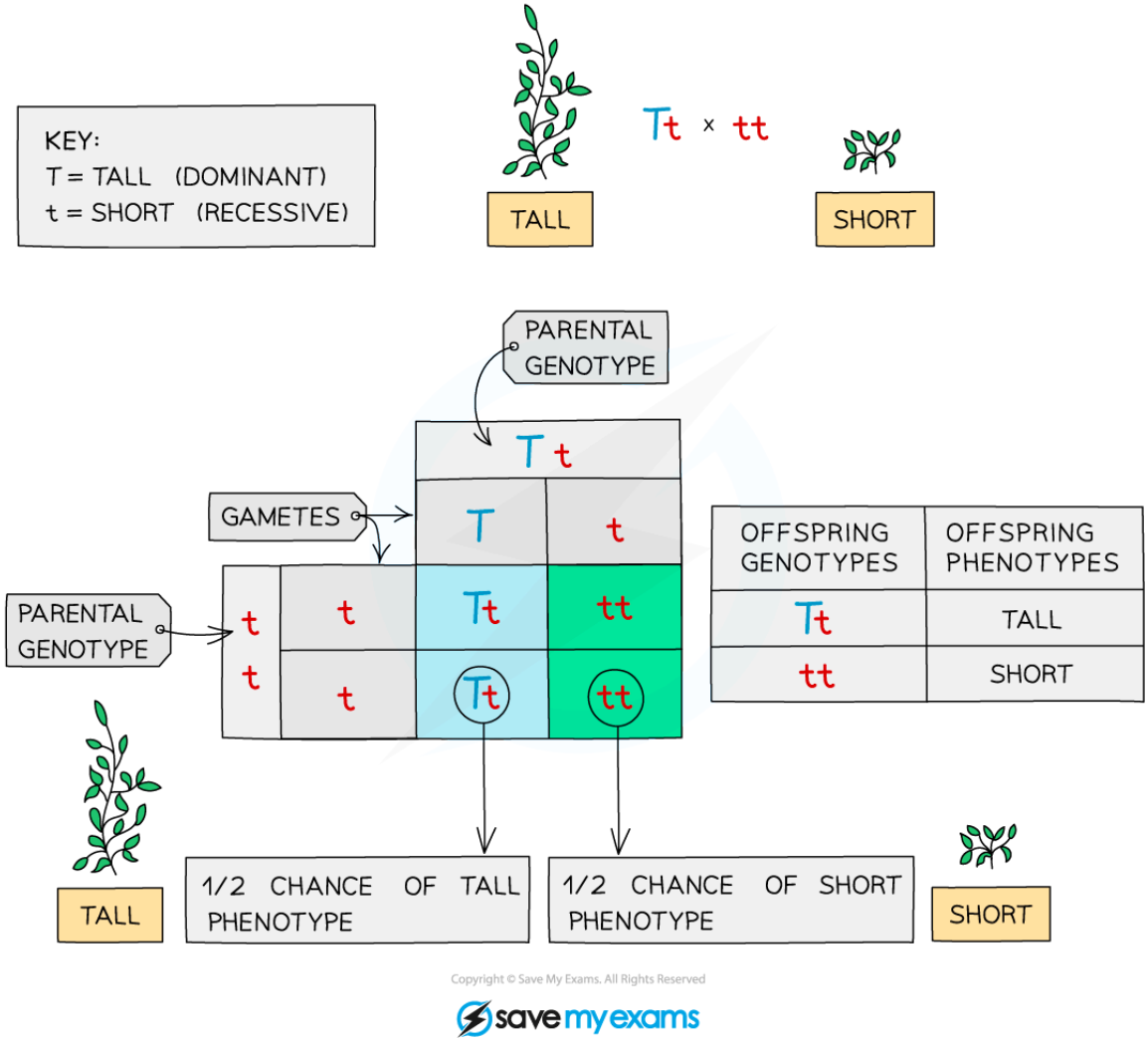
Crossing a heterozygous plant with a short plant

- The heterozygous plant will be tall with the genotype **Tt**
- The short plant is showing the recessive phenotype and so must be homozygous recessive – **tt**
- The results of this cross are as follows:



A cross between a heterozygous plant with a short plant

How to construct Punnett squares

- Determine the **parental genotypes**
- Select a letter that has a clearly different lower case, for example, Aa, Bb, Dd
- Split the alleles for each parent and add them to the Punnett square around the outside
- Fill in the middle four squares of the Punnett square to work out the **possible genetic combinations in the offspring**
- You may be asked to comment on the **ratio** of different allele combinations in the offspring, **calculate percentage chances** of offspring showing a specific characteristic or to determine the phenotypes of the offspring
- Completing a Punnett square allows you to predict the probability of different outcomes from monohybrid crosses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Calculating probabilities from Punnett squares

- A Punnett square diagram shows the **possible combinations of alleles** that could be produced in the offspring
- From this, the **ratio** of these combinations can be worked out
- However, you can also make **predictions** of the offsprings' characteristics by calculating the **probabilities** of the different phenotypes that could occur
  - For example, in the second genetic cross (F2 generation) that was given earlier (see above), two plants with the genotype **Tt** (heterozygous) were bred together
  - The possible combinations of offspring bred from these two parent plants are: **TT** (tall), **Tt** (tall), **tt** (short
  - The offspring penotypes showed a **3:1** ratio of tall : short
  - Using this ratio, we can calculate the probabilities of the offspring phenotypes
  - The probability of an offspring being tall is 75%
  - The probability of an offspring being short is 25%

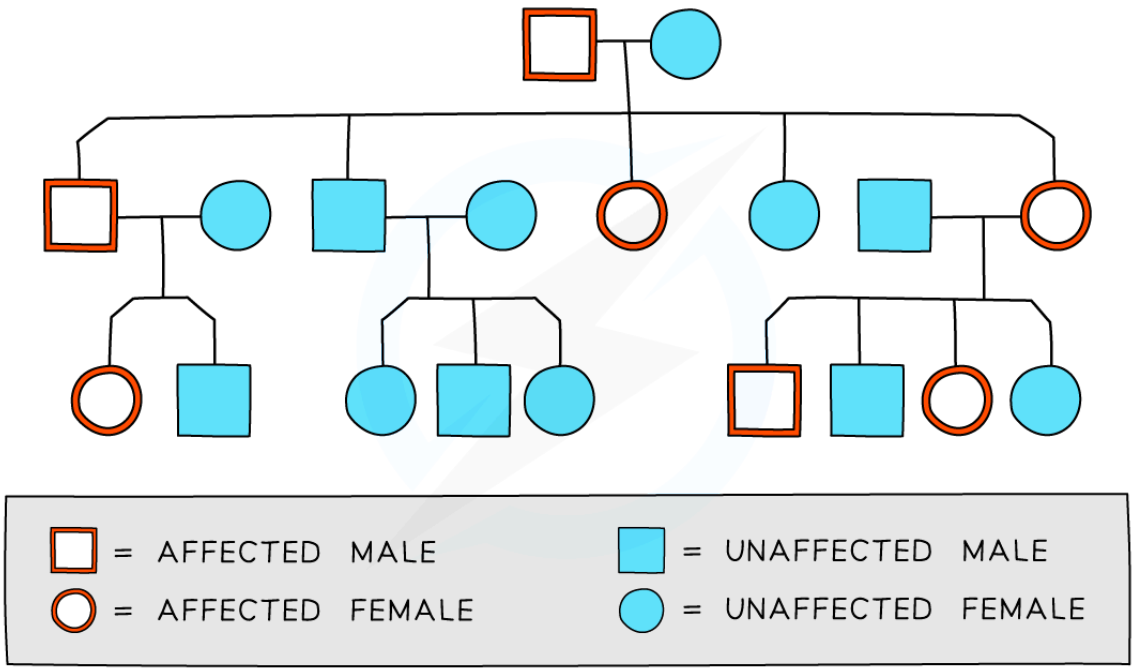
Examiner Tips and Tricks

If you are asked to use your own letters to represent the alleles in a Punnett square, try to choose a letter that is obviously different as a capital than the lower case so the examiner is not left in any doubt as to which is dominant and which is recessive.

Your notes

Family Pedigrees

- Family pedigree diagrams are usually used to trace the **pattern of inheritance** of a specific characteristic (usually a disease) **through generations of a family**
- This can be used to work out the probability that someone in the family will inherit the **genetic disorder**



A family pedigree chart

- Males are indicated by the **square shape** and females are represented by **circles**
- Affected individuals are **red** and unaffected are **blue**
- **Horizontal lines** between males and females show that they have **produced children** (which are shown underneath each couple)
- The family pedigree above shows:

- Both males and females are affected
- Every generation has affected individuals
- That there is one family group that has no affected parents or children
- The other two families have one affected parent and affected children as well


Your notes

Your notes

Inheritance of Sex

- Sex is determined by an **entire chromosome pair** (as opposed to most other characteristics that are just determined by one or a number of genes)
- **Females** have the sex chromosomes **XX**
- **Males** have the sex chromosomes **XY**
- As only a father can pass on a Y chromosome, he is **responsible for determining the sex of the child**

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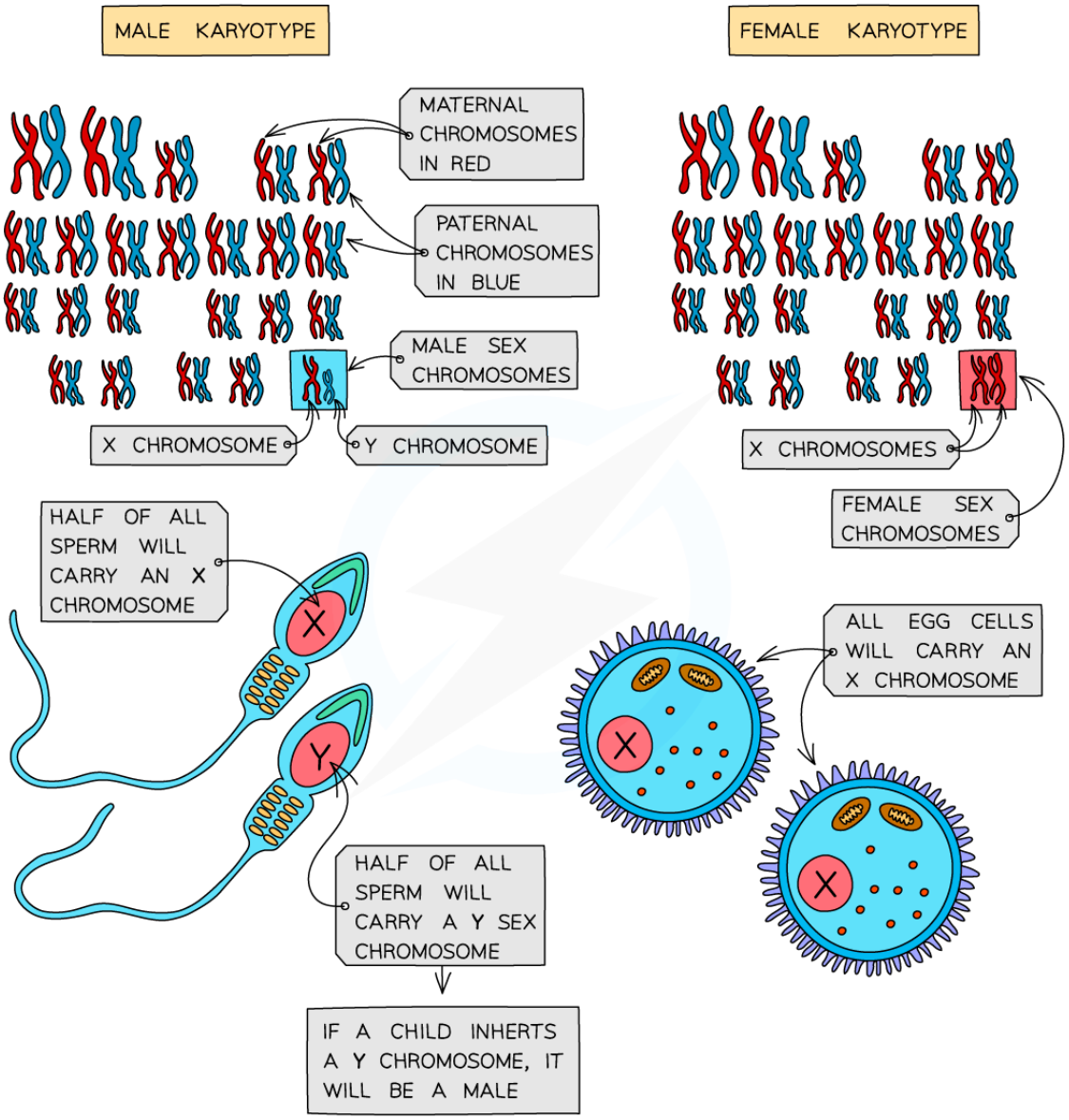
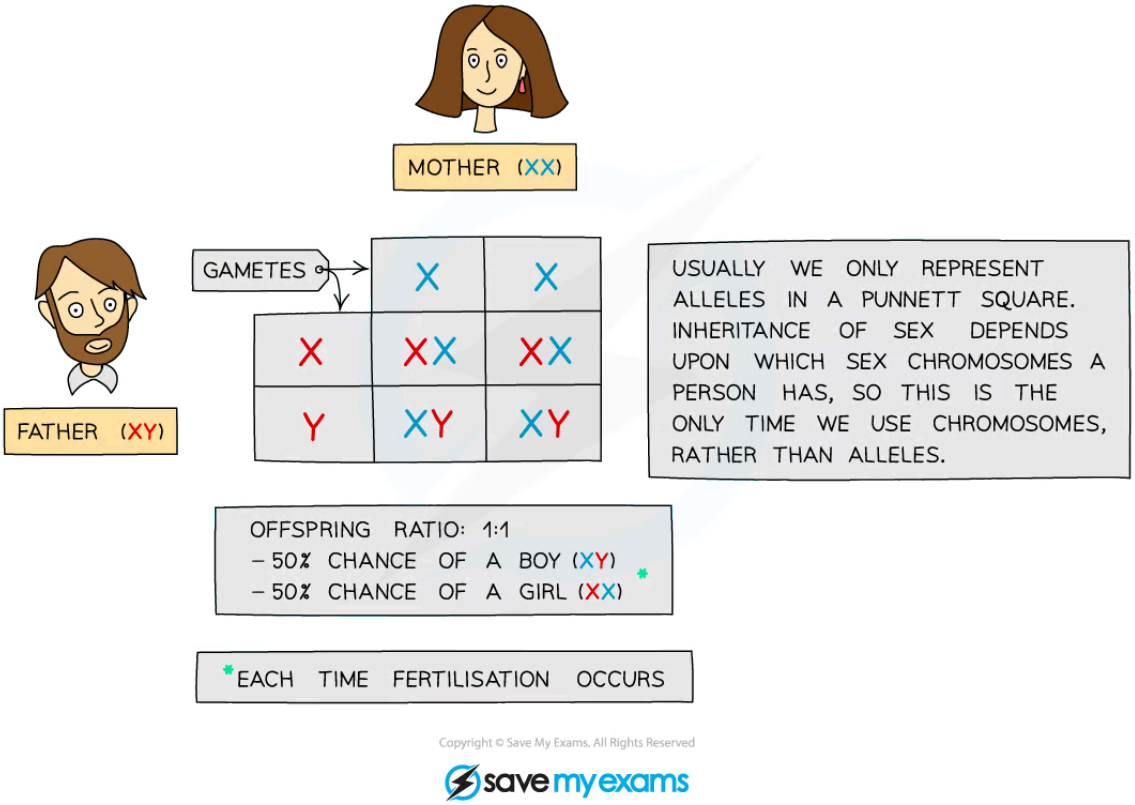


Sperm cells determine the sex of offspring

- The inheritance of sex can be shown using a **genetic diagram** (known as a **Punnett square**), with the X and Y chromosomes taking the place of the alleles usually written in the boxes

Your notes



Punnett square showing the inheritance of sex

Codominance

Your notes

Codominance

- On occasion, **both alleles within a genotype are expressed** in the phenotype of an individual - this is known as **codominance**
- Inheritance of blood group is an example of **codominance**
- There are **three** alleles of the gene governing this instead of the usual two
  - I represents the **gene** and the superscript A, B and O represent the **alleles**
- Alleles I^A and I^B are **codominant**, but both are dominant to I^O
- I^A results in the production of **antigen A** in the blood
- I^B results in the production of **antigen B** in the blood
- I^O results in **no antigens** being produced in the blood
- These three possible alleles can give us the following genotypes and phenotypes

Blood Phenotypes Table

GENOTYPE	PHENOTYPE
$I^A I^A$ OR $I^A I^O$	A
$I^B I^B$ OR $I^B I^O$	B
$I^A I^B$	AB
$I^O I^O$	O

- We can use genetic diagrams to predict the outcome of crosses that involve codominant alleles:

'Show how a parent with blood group A and a parent with blood group B can produce offspring with blood group O'

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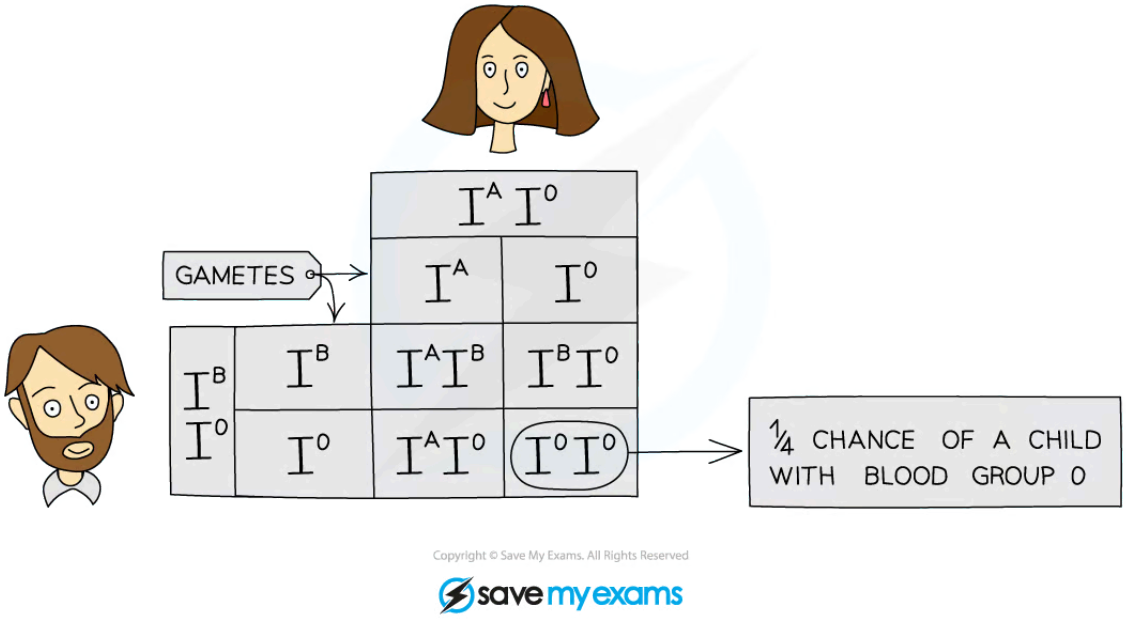


Punnett square showing the inheritance of Blood Group

- The parent with blood group A has the genotype I^AI^O
- The parent with the blood group B has the genotype I^BI^O
- We know these are their genotypes (as opposed to both being homozygous) as they are able to produce a child with blood group O and so the child must have inherited an allele for group O from each parent
- Parents with these blood types have a 25% chance of producing a child with blood type O

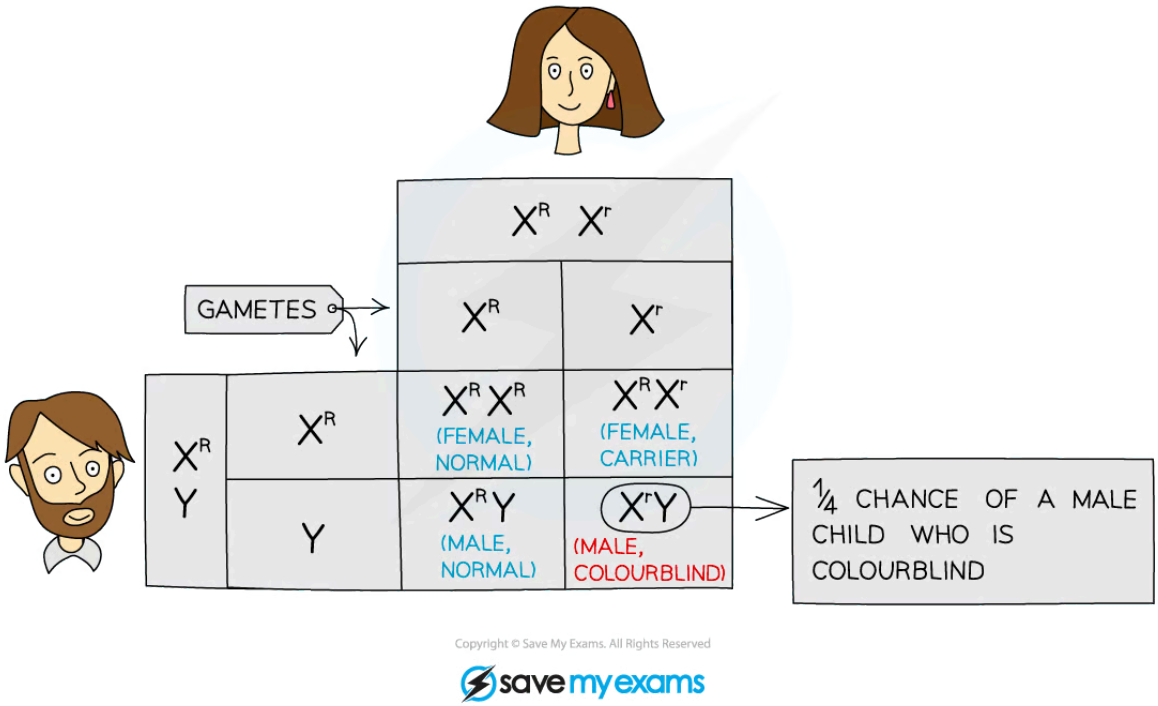
Your notes

Sex-Linked Characteristics

Higher tier only

- When alleles that control a particular characteristic are found on the **sex chromosomes**, we describe the inheritance that results as **'sex linked'**
- In almost all cases, there are **only alleles on the X chromosome** as the **Y chromosome is much smaller**
- Because males **only have one X chromosome**, they are **much more likely to show sex-linked recessive conditions** (such as red-green colour blindness and haemophilia)
- Females, having two copies of the X chromosome, are likely to inherit one dominant allele that masks the effect of the recessive allele
- A female with one recessive allele masked in this way is known as a **carrier**; she doesn't have the disease, but she has a 50% chance of passing it on to her offspring
- If that offspring is a male, he will have the disease
- The results of a cross between a normal male and a female who is a carrier for colourblindness is as follows:

Your notes



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

- In the cross above, there is a 25% chance of producing a male who is colourblind, a 25% chance of producing a female carrier, a 25% chance of producing a normal female and a 25% chance of producing a normal male

Your notes

Polygenic Inheritance

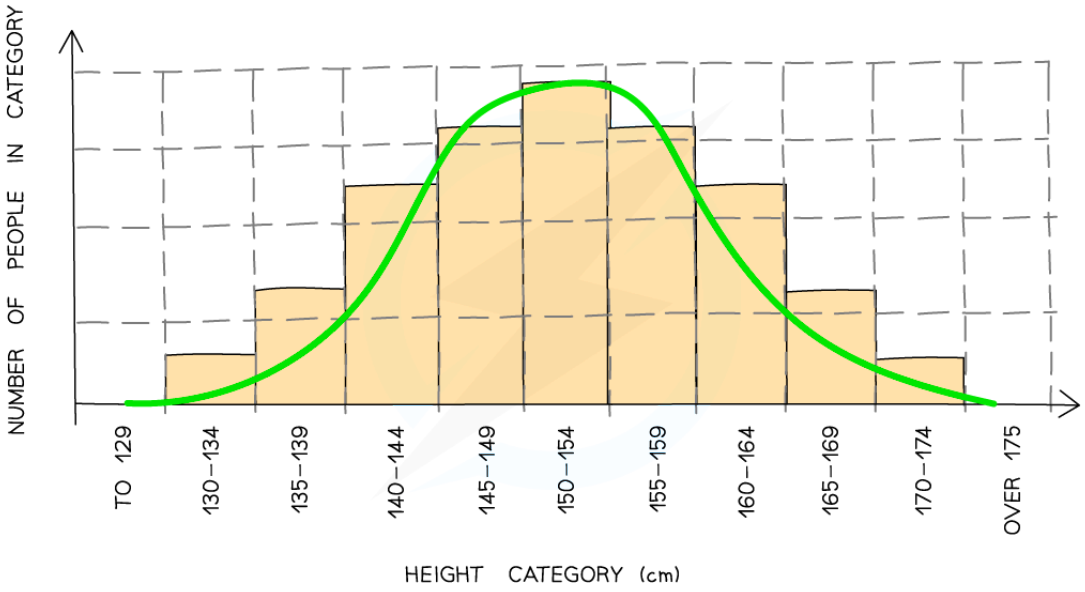
- Most characteristics are a result of multiple genes interacting, rather than a single gene
- Characteristics that are controlled by **more than one gene** are described as being **polygenic**
- Polygenic characteristics have phenotypes that can show a wide range of combinations in features
- The inheritance of these polygenic characteristics is called **polygenic inheritance** (poly = many/more than one)
- Polygenic inheritance is difficult to show using genetic diagrams because of the wide range of combinations
- An example of polygenic inheritance is **eye colour** – while it is true that brown eyes are dominant to blue eyes, it is not as simple as this as eye colour is controlled by several genes
- This means that there are several different phenotypes beyond brown and blue; green and hazel being two examples

Variation

Your notes

Types of Variation

- Variation is defined as **differences between individuals of the same species**
- Variation can be divided into two types depending on **how you are able to group the measurements:**
  - **Continuous variation** is when there are very many small degrees of difference for a particular characteristic between individuals and they are arranged in order and can usually be measured on a scale
    - Examples include height, mass, finger length etc. where there can be many 'in-between groups
  - **Discontinuous variation** is when there are distinct differences for a characteristic
    - For example, people are either blood group A, B, AB or O; are either male or female; can either roll their tongue or not - there are no 'in-betweens'
- When graphs of these data are plotted, continuous variation gives smooth bell curves (a result of all the small degrees of difference), whereas discontinuous gives a 'step-like' shape



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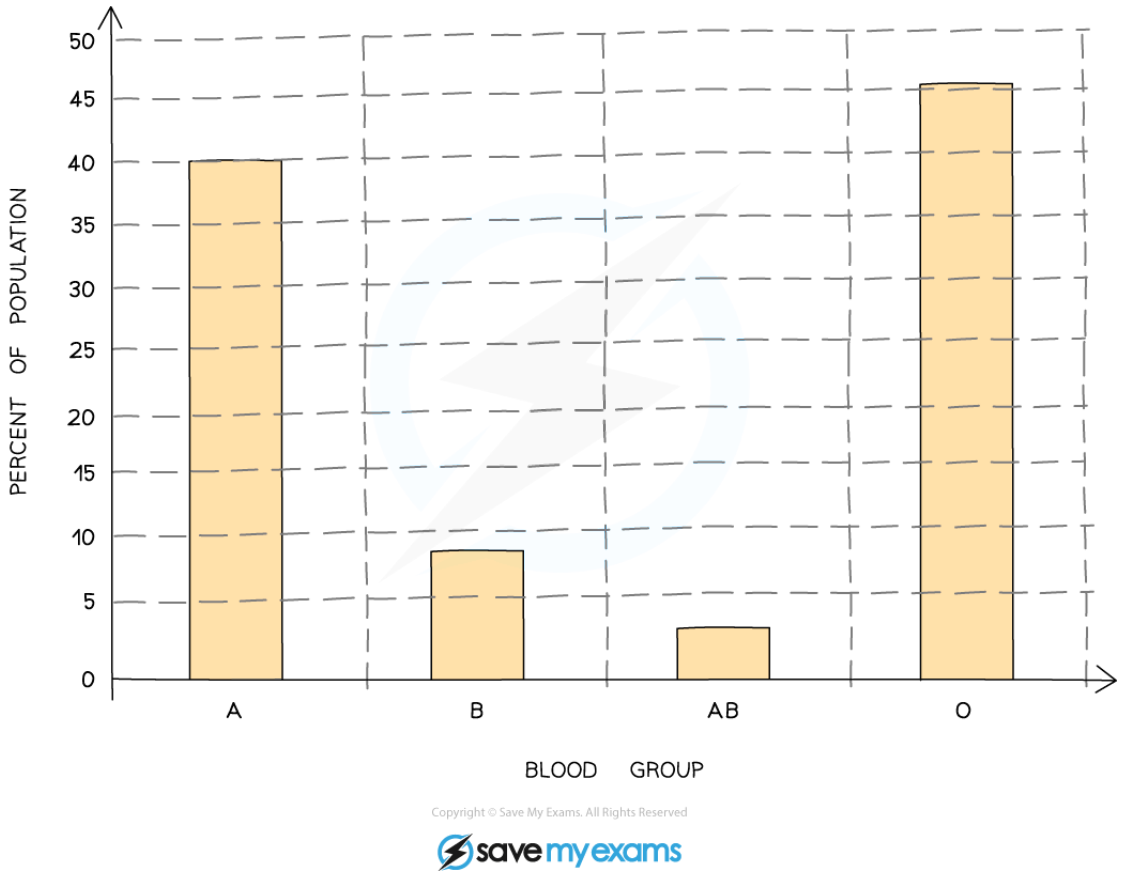
Height is an example of continuous variation which gives rise to a smooth bell-shaped curve when plotted as a graph



Blood group is an example of discontinuous variation which gives rise to a step-shaped graph

Phenotypic Variation

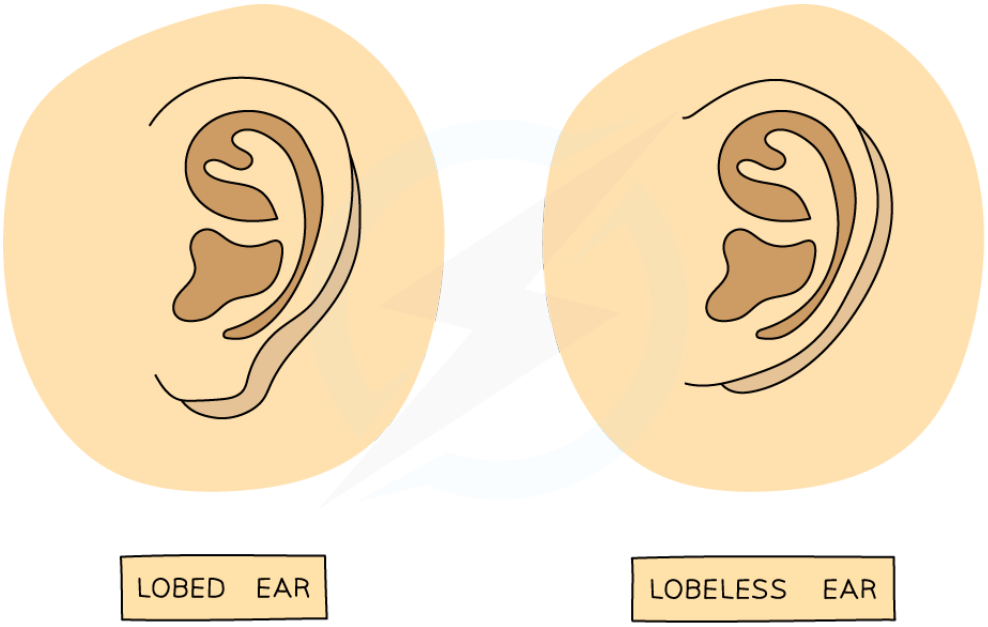
- Phenotypic variation can be caused in two main ways:
  - It can be **genetic** - controlled entirely by genes
  - Or it can be **environmental** - caused entirely by the environment in which the organism lives

Genetic variation

- Meiosis creates **genetic** variation **between the gametes** produced by an individual
- This means each gamete carries substantially **different alleles**

Your notes

- During fertilization, any male gamete can fuse with any female gamete to form a zygote
- This **random fusion of gametes at fertilization creates genetic variation between zygotes** as each will have a unique combination of alleles
- Zygotes eventually grow and develop into adults
- Examples of genetic variation in humans include:
  - Blood group
  - Eye colour
  - Gender
  - Ability to roll tongue
  - Whether ear lobes are free or fixed



Whether earlobes are attached (lobeless) or free (lobed) is an example of genetic variation

Environmental variation

- Characteristics of all species can be affected by environmental factors such as climate, diet, accidents, culture and lifestyle

- In this instance 'environmental' simply means 'outside of the organism' and so can include factors like climate, diet, culture, lifestyle and accidents during lifetime
- Examples include:
  - An accident may lead to **scarring** on the body
  - Eating too much and not leading an active lifestyle will cause **weight gain**
  - Being raised in a certain country will cause you to speak a certain **language** with a certain **accent**
  - A plant in the shade of a big tree will grow **taller** to reach more light


Your notes

Genetic and environmental causes

- Discontinuous variation is usually caused by **genetic variation alone**
- Continuous features often vary because of a **combination of genetic and environmental** causes, for example:
  - Tall parents will **pass genes** to their children for height
  - Their children have the **genetic potential** to also be tall
  - However if their **diet is poor** then they will not grow very well
  - Therefore their **environment** also has an impact on their height
- Another way of looking at this is that although genes decide what characteristics we inherit, the surrounding environment will affect how these inherited characteristics develop

Mutations

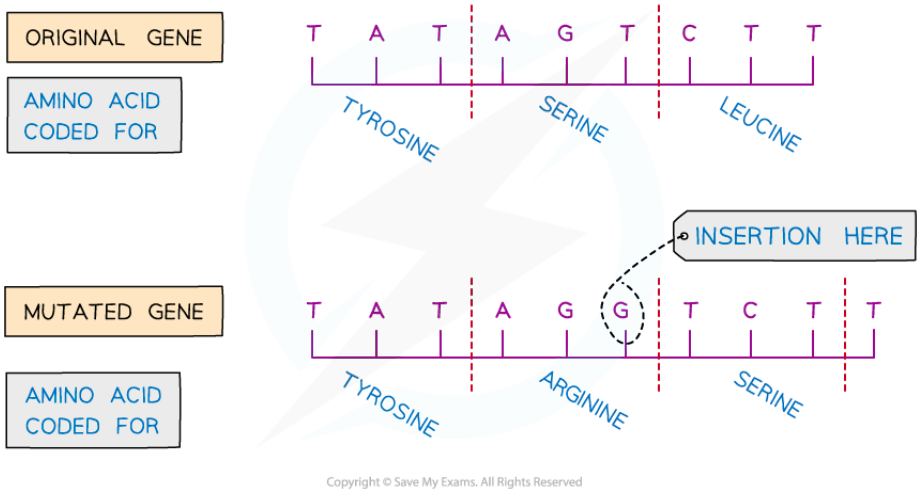
Your notes

Mutations

- Mutations are **rare, random changes** that occur in the **sequence of DNA bases** in a gene or a chromosome
- Mutations occur **continuously**
- As the DNA base sequence determines the sequence of amino acids that make up a protein, **mutations in a gene can sometimes lead to a change in the protein that the gene codes for**
- **Most mutations do not alter the protein** or only alter it slightly so that its appearance or function is not changed
- There are different ways that a mutation in the DNA base sequence can occur

Insertions

- **A new base is randomly inserted** into the DNA sequence
- An insertion mutation **changes the amino acid that would have been coded for** by the group of three bases in which the mutation occurs
  - Remember – every group of three bases in a DNA sequence codes for an amino acid
- An insertion mutation also has a **knock-on effect** by **changing the groups of three bases further on in the DNA sequence**



An example of an insertion mutation

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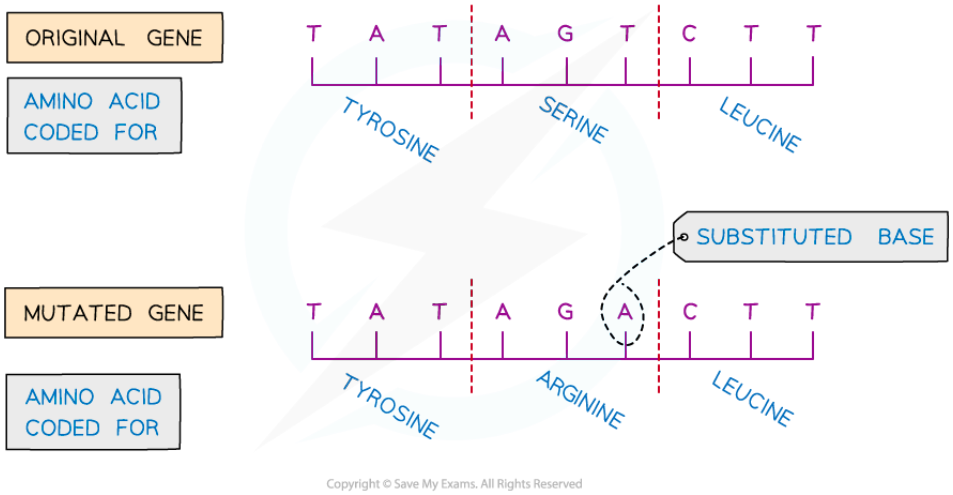
Deletions

- **A base is randomly deleted** from the DNA sequence
- Like an insertion mutation, a deletion mutation **changes the amino acid that would have been coded for** by the group of three bases in which the mutation occurs
- Like an insertion mutation, a deletion mutation also has a **knock-on effect** by **changing the groups of three bases further on in the DNA sequence**

Substitutions

- **A base in the DNA sequence is randomly swapped for a different base**
- **Unlike** an insertion or deletion mutation, a substitution mutation will **only change the amino acid for the group of three bases in which the mutation occurs**; it will **not have a knock-on effect**



An example of a substitution mutation

The Effects of Mutations

- **Most mutations do not alter the protein** or only alter it slightly so that its appearance or function is not changed
- However, a **small number of mutations** code for a **significantly altered protein with a different shape**
- This **may affect the ability of the protein to perform its function**. For example:

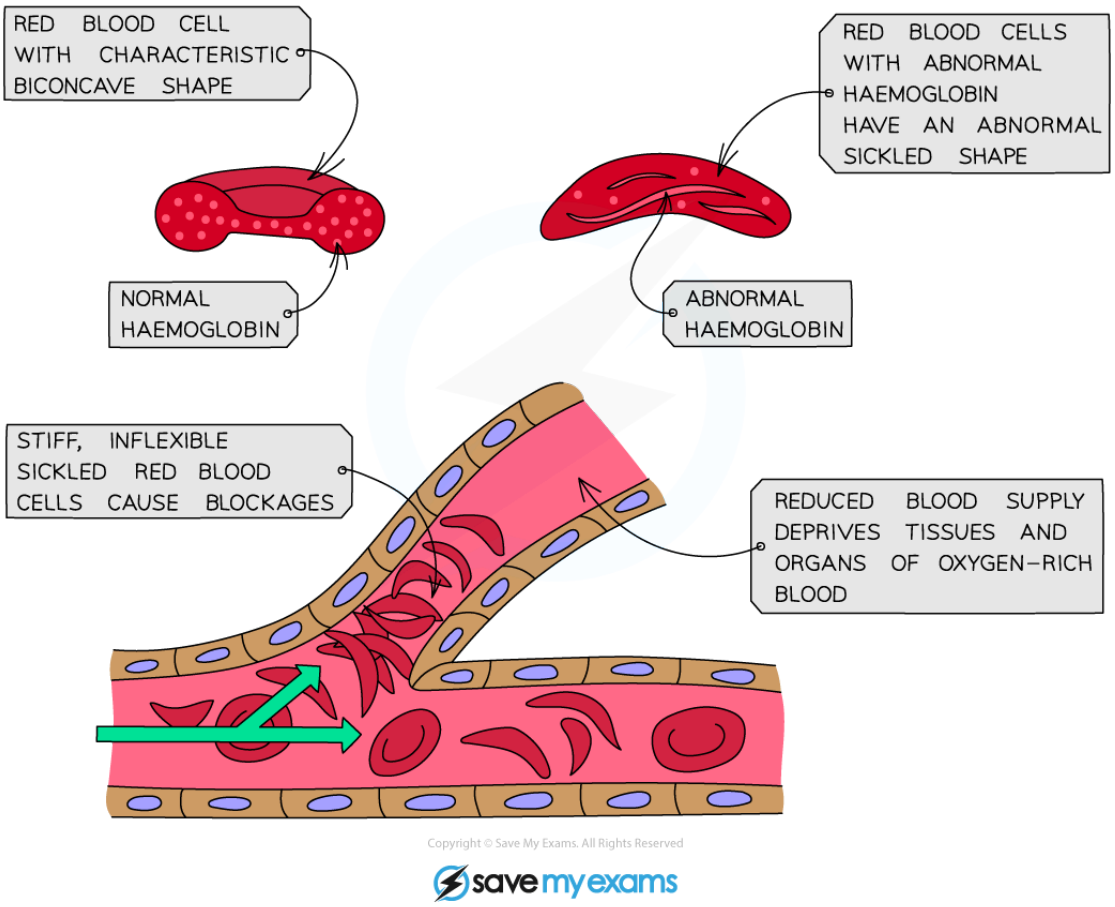

Your notes

- If the shape of the active site on an enzyme changes, the substrate may no longer be able to bind to the active site
- A structural protein (like collagen) may lose its strength if its shape changes

- On rare occasions mutations lead to the development of new alleles and so new phenotypes
- Occasionally, the new allele (and its resulting phenotype) gives the individual a **survival advantage** over other members of the species
- For example:
  - A bird develops a mutation leading to a change in feather colours
  - This makes it more attractive to birds of the opposite sex
  - Which causes the bird to breed more frequently and have more chances of passing on the mutated phenotype to the next generation
- Mutations can also lead to **harmful changes** that can have dramatic effects on the body - for example, **sickle cell anaemia** in humans

Sickle cell anaemia

- Sickle cell anaemia was the first genetic disease to be described in terms of a **gene mutation**
- A gene mutation is a **change in the base sequence of DNA**
- The mutation **changes the molecule haemoglobin**, causing the red blood cells (RBC's) to become stiff and sometimes **sickle-shaped** when they release oxygen to the body tissues
- The sickled cells tend to get **stuck** in narrow blood vessels, **blocking the flow of blood**
- As a result, those with sickle cell disease suffer painful "crises" in their joints and bones
- They may suffer **strokes, blindness, or damage to the lungs, kidneys, or heart**. They must often be hospitalized for blood transfusions and are at risk for a life-threatening complication called **acute chest syndrome**
- Although many sufferers of sickle cell disease die before the age of 20, modern medical treatments can sometimes prolong these individuals' lives into their 40s and 50s



Your notes

Sickle cell anaemia is caused by abnormal haemoglobin which changes the shape of red blood cells

The Causes of Mutations

- Mutations happen **spontaneously and continuously** but their frequency can be increased by exposure to the following:
  - **Gamma rays, x – rays and ultraviolet rays** - all types of ionising radiation which can damage bonds and cause changes in base sequences
  - **Certain types of chemicals** - for example chemicals such as tar in tobacco
- Increased rates of mutation can cause cells to become **cancerous**, which is why the above are linked to increased incidence of different types of cancer