SL IB Biology

Your notes

Inheritance

Contents

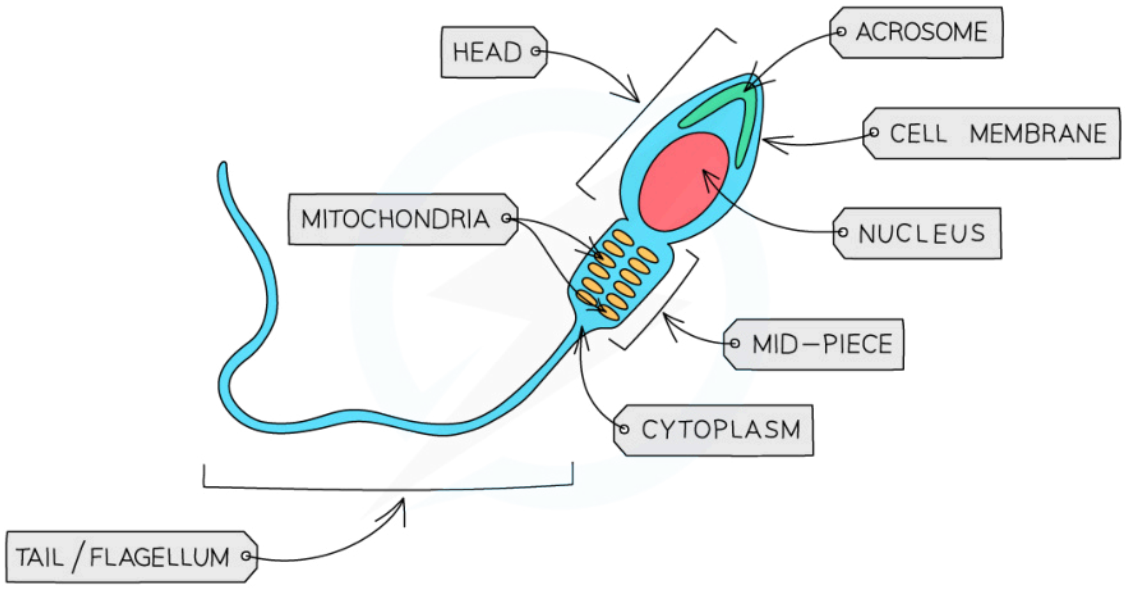
Genetic Inheritance & Genetic Crossing

Your notes

Inheritance: Gametes & Fertilisation

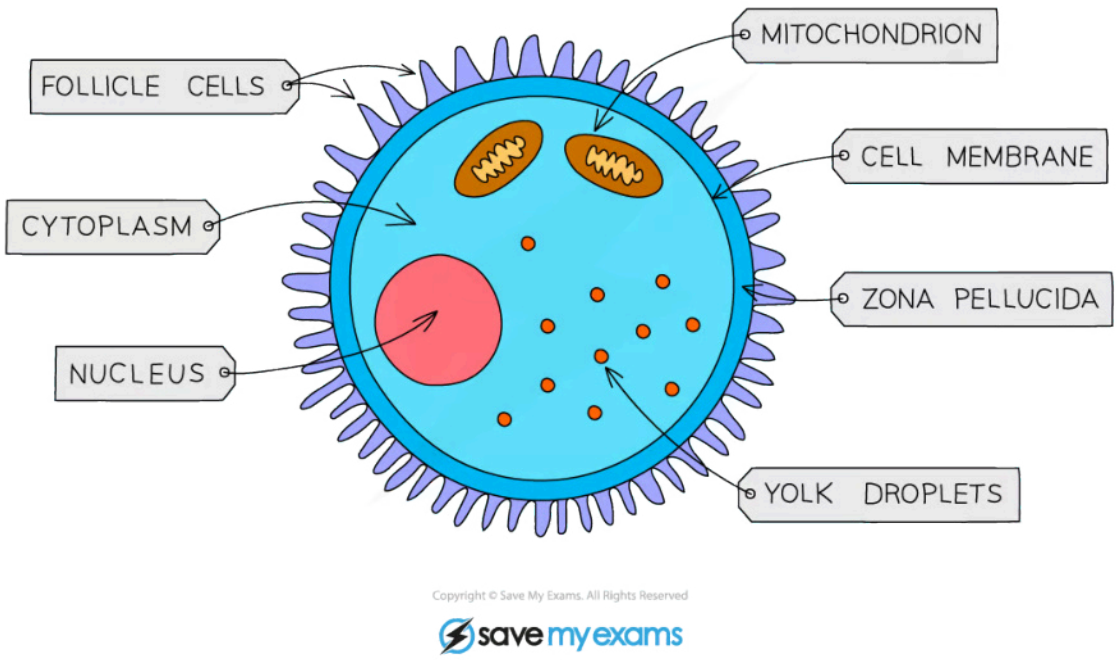
- Gametes are the **sex cells** of an organism
- For example, the **sperm** and **egg** (ovum) cells in humans
  - The **egg is larger than the sperm** as most of its space contains food to nourish a growing embryo
  - The sperm cell contains **many mitochondria** to release energy for its motion
- Gametes **fuse** during fertilization to form a **zygote** (fertilised egg cell)
- These sex cells are formed during **meiosis** and only have **one** copy of each chromosome and so are **haploid** cells
  - For humans, that means the sperm and egg cells contain **23 single chromosomes** in their nucleus (as opposed to diploid cells which contain 46 chromosomes, or 23 pairs)
  - As there is only **one chromosome from each homologous pair** there is only **one allele** of each gene present
    - This allele may be dominant, recessive or **co-dominant**

Sperm cell diagram



Egg cell diagram

Your notes



The structure of human gametes – the sperm and egg

- Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele **or** different alleles
- Sexual reproduction is **a process involving the fusion of the nuclei of two gametes (sex cells) to form a zygote** (fertilised egg cell) and the production of offspring that are **genetically different from each other**
- Fertilisation is defined as the **fusion of gamete nuclei**, and as each gamete comes from a different parent, there is **variation** in the offspring
- When a male and female gamete fuse their **chromosomes are combined**
- This means the resulting zygote is **diploid**
  - The zygote contains two chromosomes of each type
- It will therefore have **two alleles** of each gene
  - If the two alleles for a particular gene are the **same** then the genotype is described as **homozygous**
  - If the two alleles for a particular gene are **different** then the genotype is described as **heterozygous**

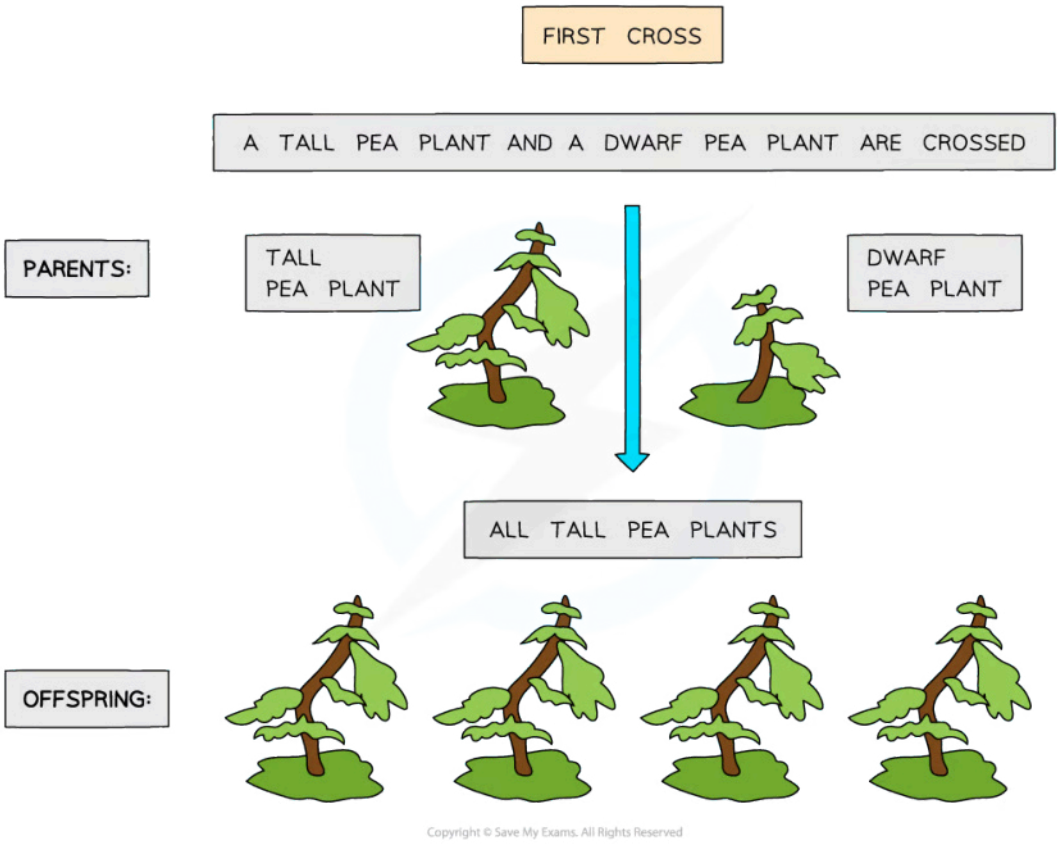
Genetic Crosses in Flowering Plants

Your notes

- **Gregor Mendel** was an Austrian monk
- In the mid-19th century, Mendel carried out **breeding experiments** on large numbers of pea plants whilst looking after the monastery gardens
- He studied how **characteristics were passed on between generations of plants**
- Due to his extensive work on the understanding of inheritance, he is sometimes called the **Father of Genetics**
- Mendel carefully **transferred pollen** from one pea plant to the reproductive parts of another
  - **Pollen contains the male gamete** and is located on the anther of the flower
  - The female gametes are located in the **ovary**
  - The plants reproduce sexually and require **pollination** for fertilisation
  - This technique **eliminated any uncertainty** from his data since he knew which pollen had fertilised each of the plants
- He **collected the pea seeds from these plants** and grew them in favourable conditions to find out their characteristics
- He also **cross-bred offspring peas** in order to find out which, if any characteristics would appear in future generations
- Mendel investigated the **height** of pea plants, the **colours of their flowers** and the **smoothness of their seed coat**

Mendel's breeding experiments of pea plants diagram

Your notes



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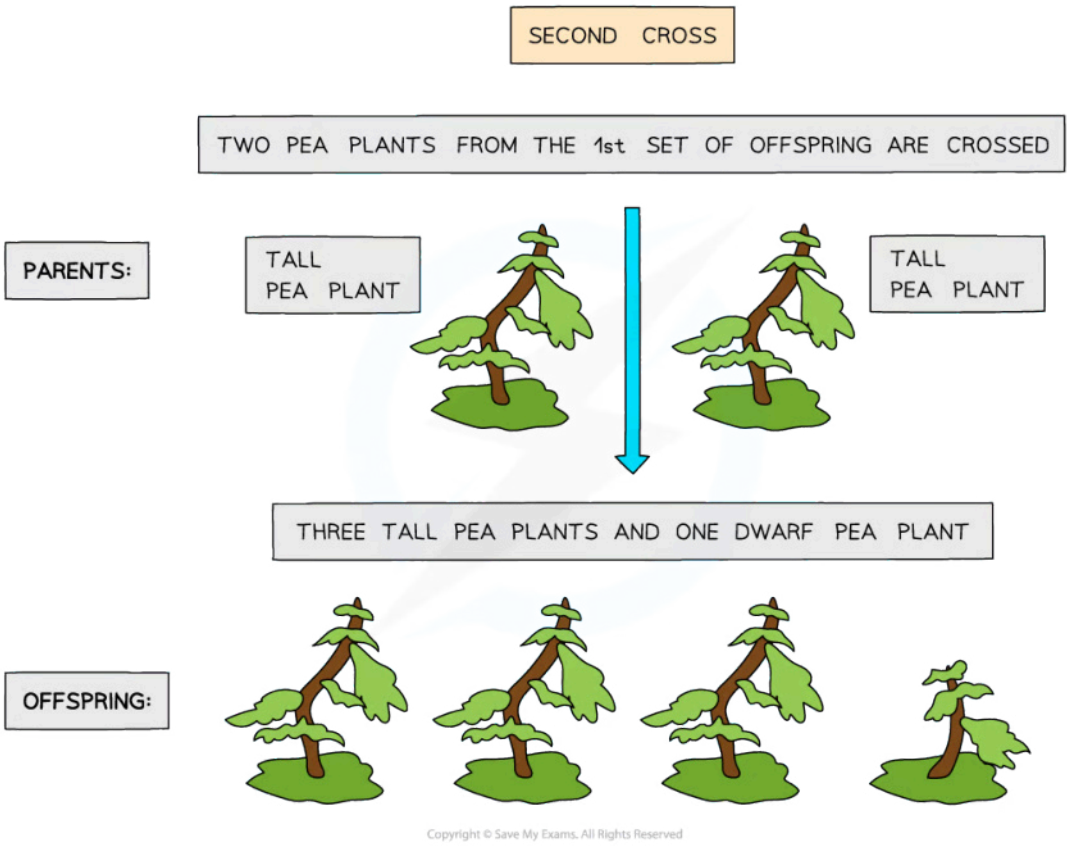


Mendel's pea plant crosses

Mendel's Pea Plant Results Table

Parental characteristics	Characteristics of first generation plants	Chraracteristics of second generation plants	Ratio of characteristics in second generation
Tall plant × dwarf plant	100% tall plants	868 tall plants and 277 dwarf plants	3.1 : 1
Round seed coat × wrinkled seed coat	100% Round seed coat	5474 round seed coat and 1850 wrinkled seed coat	3 : 1
Purple flowers × white flowers	100% Purple flowers	705 purple flowers and 224 white flowers	3.1 : 1

- Mendel found that characteristics were **inherited** in a **predictable pattern**
- **All** pea plants in the **first generation** had the same characteristic as **one** of the parental plants
- The offspring plants in the **second generation** had characteristics of **both** parent plants in a **3:1 ratio**
- Without knowing it, **Mendel had discovered genes**, he referred to them as **'units of inheritance'**
- He also discovered that some genes are **dominant** and some genes are **recessive**
- Different forms of the same gene are called **alleles**

- A **monohybrid trait** is one that is controlled by only **one gene**
- Generally, we consider that such a gene has **two alleles**
  - Either: one allele is dominant and the other is recessive
  - Or: the alleles are co-dominant
- A monohybrid cross starts with **pure-breeding parents** (homozygous), each displaying a different phenotype
  - This generation is known as the parental generation, denoted as the **P generation**
- The **purpose of a Punnett grid** is to predict the probability of a certain offspring displaying a certain genotype or phenotype
  - In the case where multiple offspring are produced, Punnett grids can **predict the numbers of offspring** that will display a certain genotype or phenotype after a cross

Steps in constructing a Punnett Grid

1. Write down the **parental phenotypes** and **genotypes**
2. Write down all the **possible gamete genotypes** that each parent could produce for sexual reproduction
   - A useful convention is to write the gamete genotypes **inside a circle** to denote them as gametes (haploid cells)
3. Place each parental genotype **against one axis** of a Punnett grid (2 x 2 table)
4. In the boxes of the Punnett grid, combine the gametes into the possible genotypes of the offspring
   - This gives the offspring of the **F_1 generation** (1^{st} filial generation)
5. List the **phenotype** and **genotype ratios** for the offspring

Worked example

Sweet peas grow pods that are either green or yellow. The allele for green, **G**, is dominant to the allele for yellow, **g**. Construct a Punnett grid to predict the outcome when crossing green and yellow pure-bred plants to show the F_1 generation offspring. Using plants from the F_1 generation, construct a second Punnett grid to show the outcomes of the F_2 generation.

Step 1: Write down the parental phenotype and genotypes

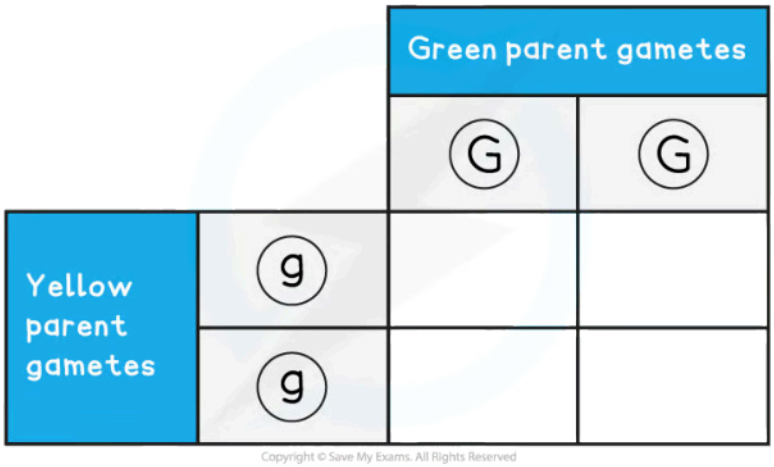
Green coloured pods	Yellow coloured pods
GG	gg

Step 2: Write down all the possible gamete genotypes that each parent could produce

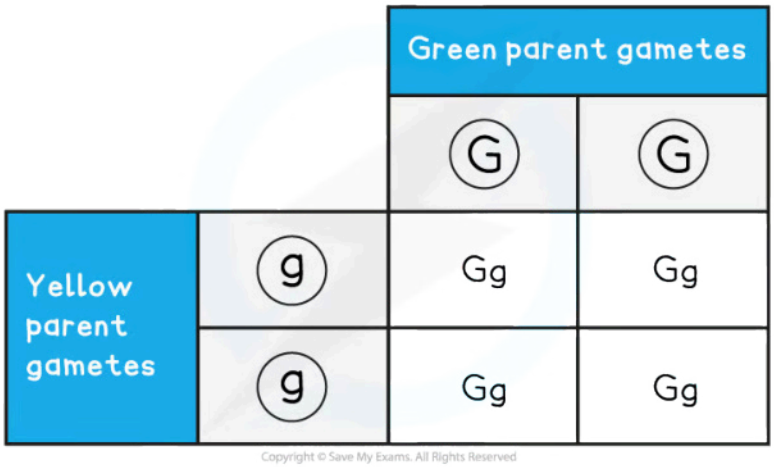
Your notes

Your notes

Step 3: Place each parental genotype against one axis of a Punnett grid (2 x 2 table)



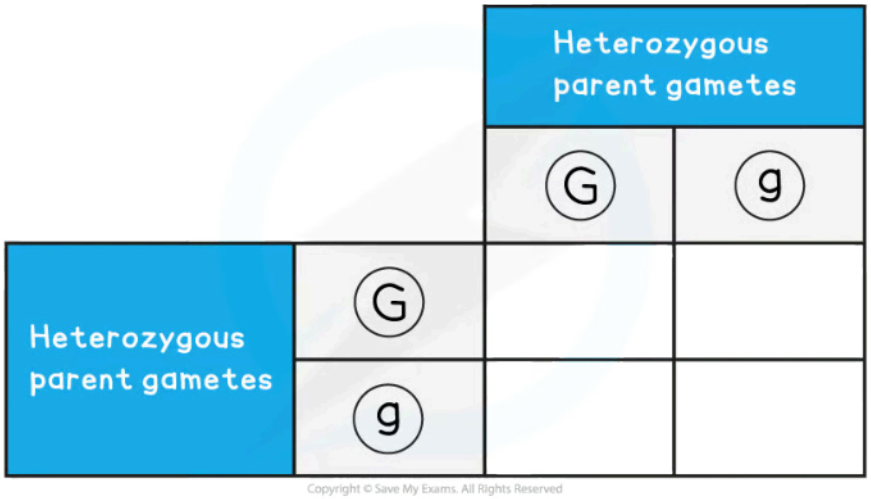
Step 4: Combine the gametes in each box of the Punnett grid



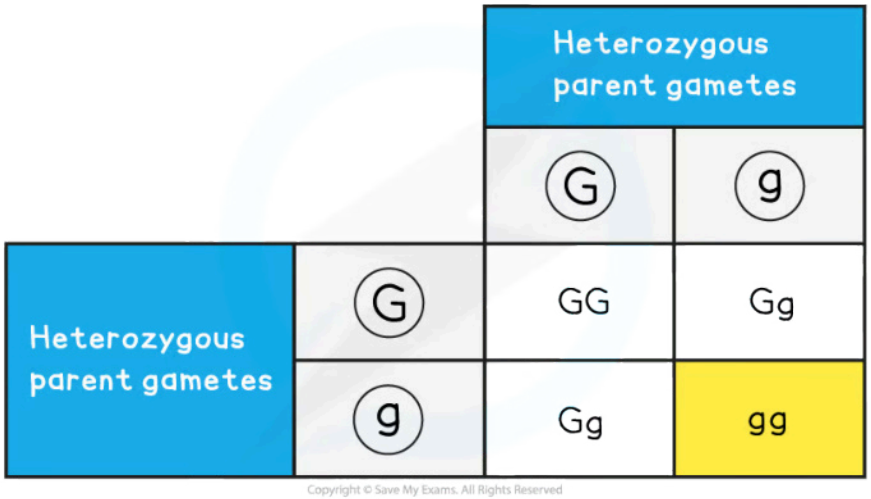
Genotypes of the F1 cross between homozygous green (GG) and homozygous yellow (gg) pea plants. All offspring (100%) have the genotype Gg and the phenotype is green.

Step 5: Take two heterozygous offspring from the F_1 generation and cross them

Your notes



Step 6: Combine the gametes in each box of the Punnett grid



Punnett grid showing the results of the F2 generation

Phenotype ratio is 3:1 green:yellow, Genotype ratio is 1 GG: 2 Gg: 1 gg

- Plants can sexually reproduce in different ways:
  - Some plants have the male and female reproductive parts within the **same flower**
  - Others have male flowers and female flowers on the **same plant**
  - Others have **different male and female plants**
- Plants with male and female reproductive parts on the same plant can be capable of **self-pollination and self-fertilisation**
- Farmers and ornamental plant growers can control the way their plants reproduce by **artificially pollinating** them

- If a grower thinks a trait is useful or profitable they may choose to **self-pollinate** the favoured plants to keep the desirable traits in the next generation
- Growers can also **cross-pollinate** by artificial pollination between different plants with favoured traits, with the goal to create new generation of plants will possess the desirable traits from both parent plants
- **Genetic crosses** can be used to **predict** and plan for these outcomes


Your notes

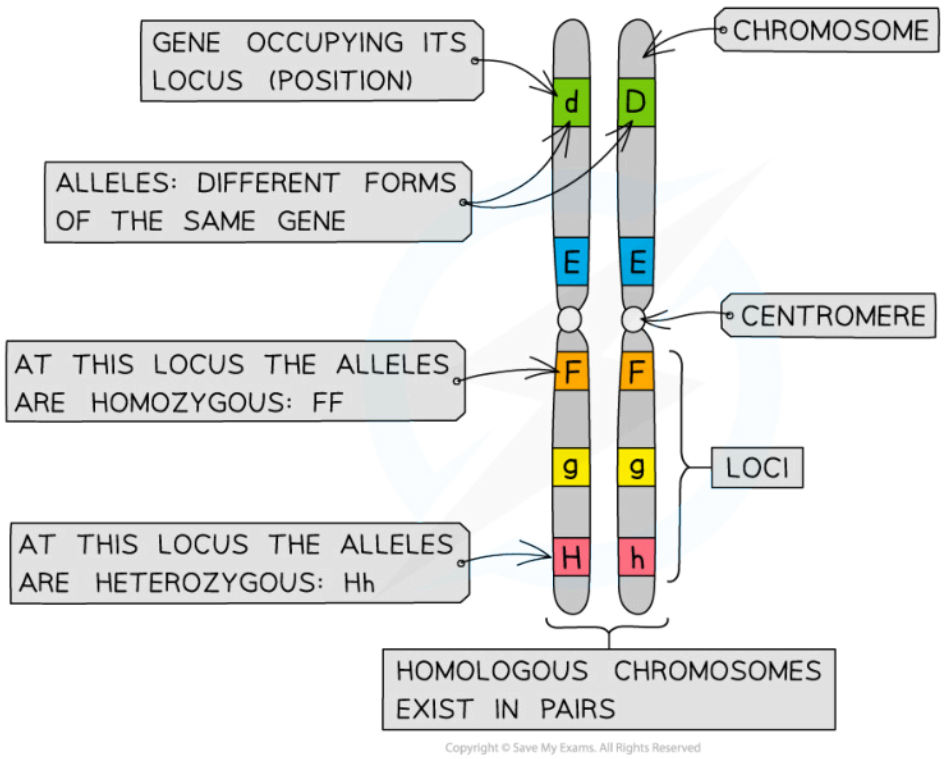
Inheritance: Terminology

Your notes

Genotype

- A **gene** is a short length of DNA found on a chromosome that codes for a particular **characteristic** (by coding for the production of a specific protein)
- **Alleles** are variations of the same gene
  - As we have two copies of each chromosome, we have two copies of each gene and therefore every individual will have two alleles of each gene
  - One of the alleles is inherited **from the mother** and the other **from the father**
  - The alleles may be the same as each other, or they might be different. e.g. 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 combination of alleles that an individual organism inherits is its **genotype**
  - When the two alleles at a locus are the **same/identical**, an individual is said to have a **homozygous** genotype
  - When the two alleles at a locus are **different** the genotype is said to be **heterozygous**

Chromosome diagram



Chromosomes showing genes, loci and alleles

Exam Tip

Make sure to not use the words allele and gene interchangeably. They both have different definitions and need to be used correctly in your exams in order to gain marks.

Your notes

Phenotype

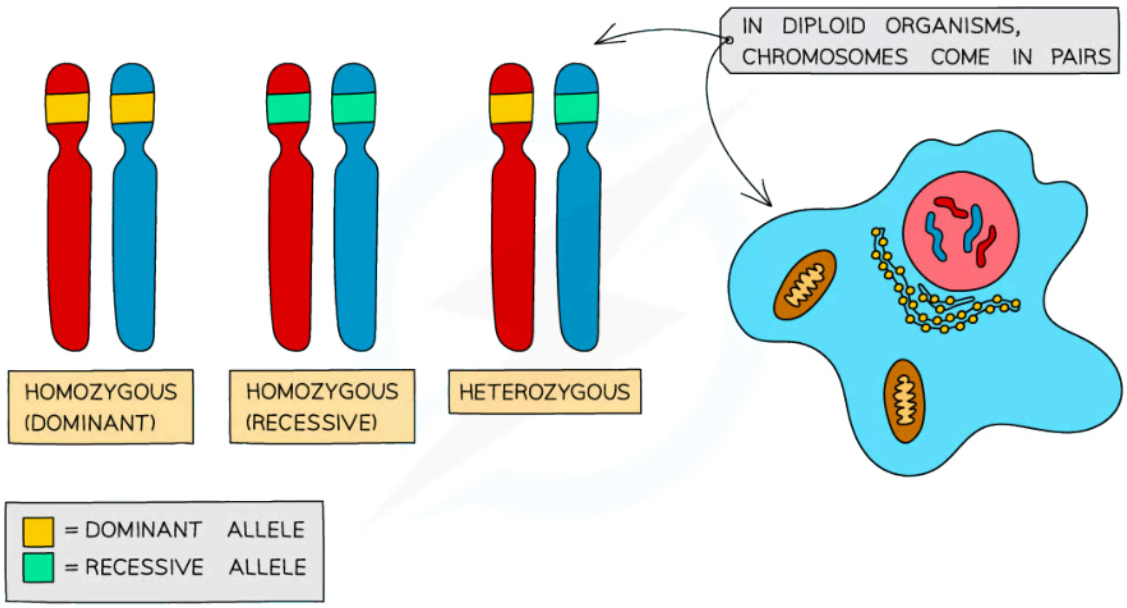
- The **observable characteristics** of an organism (seen just by looking - like eye colour, or found – like blood type) is called the **phenotype**
- The phenotype of all characteristics is determined by the following factors:
  - The **genotype** only - the combination of the two alleles for the gene for the characteristic, for example blood group is determined this way
  - The **environment** only - surroundings such as chemical or radiation exposure, diet or exercise can affect physical characteristics, for example scars and accent are determined this way
  - **Interaction** between **both** the genotype and the environment, for example height and skin colour are determined this way

Dominant & Recessive Alleles

Your notes

- Alleles can be **dominant** or **recessive**
  - A **dominant** allele **only needs to be inherited from one parent** in order for the characteristic to be expressed in the phenotype
  - A **recessive** allele needs to be **inherited from both parents in order** for the characteristic to be expressed 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)

Different forms of allele pairs diagram



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). Alleles can be dominant or recessive.

Incomplete & Codominance

Your notes

- **Co-dominant alleles** have a combined effect on the phenotype
  - The alleles are **both expressed to an equal extent** in the phenotype
- An example of codominance is in speckled chickens
  - Chickens can have different alleles for gene that determines the colour of their feathers
    - We can denote the gene for **colour** using the capital letter **C**
  - The two alleles for this gene are **white** for white feather colour, and **black** for black feather colour
    - We denote the two alleles using superscript letters, **C^W and C^B**
  - A chicken with the genotype **C^WC^W** has white feathers as their phenotype
  - A chicken with the genotype **C^BC^B** has black feathers as their phenotype
  - A chicken with the genotype **C^WC^B** has a combination of **both** feather colours, they are called **speckled colour chickens**
  - Because both alleles are expressed in the phenotype this is called **codominance**

Example of codominance in chickens diagram

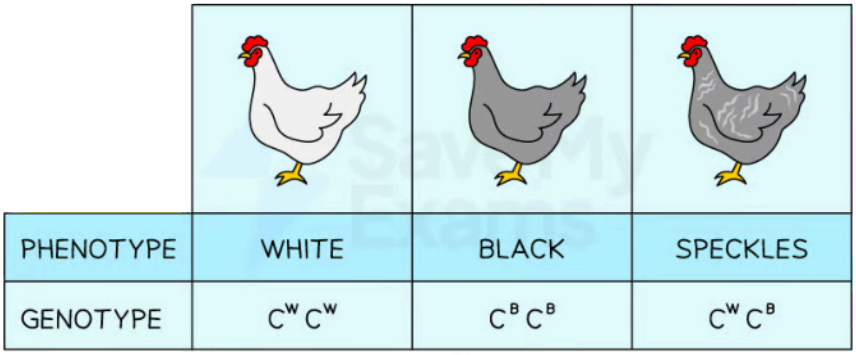


Diagram showing the phenotypes and genotypes of white, black and speckled chickens, which is an example of codominance

- **Incomplete dominance** is **similar to codominance** because two alleles are expressed together instead of just one dominant allele being expressed
- However, instead of both alleles being expressed, both alleles are **partially expressed** leading to a phenotype which is a **blend o**f both phenotypes or an **intermediate** phenotype between the two
- An example is incomplete dominance can be found in the four o'clock flower or marvel of Peru (*Mirabilis jalapa*)
  - Marvel of Peru can have different alleles for gene that determines the colour of their flowers
    - We can denote the gene for **colour** using the capital letter **C**
  - The two alleles for this gene are **white** for white flower colour, and **red** for red flower colour
    - We denote the two alleles using superscript letters, **C^W and C^R**
  - A plant with the genotype **C^WC^W** has white flowers as their phenotype
  - A plant with the genotype **C^RC^R** has red flowers as their phenotype
  - A plant with the genotype **C^WC^R** has a blend of both colours, which is expressed in the phenotype as a **pink flower colour**

- Because the flowers are neither white or red, but an **intermediate** between this two, this is **incomplete dominance**

Your notes

Example of incomplete dominance in marvel of Peru diagram

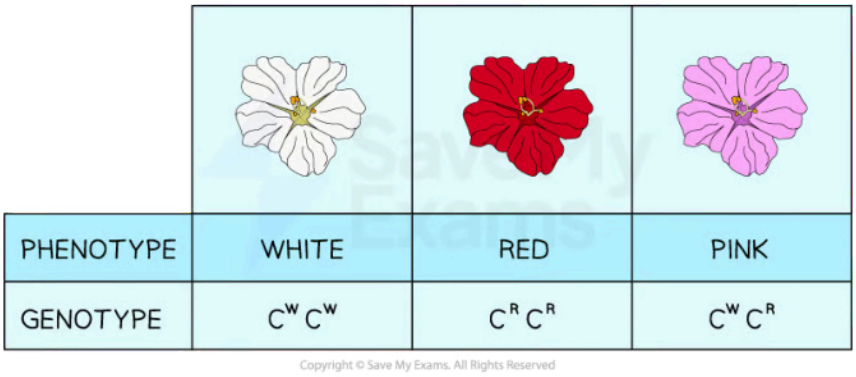


Diagram showing the phenotypes and genotypes of white, red and pink flowers of the marvel of Peru, which is an example of incomplete dominance

Exam Tip

When referring to different species examples in an exam you can use either the common name or the scientific name to gain marks. For example you could say 'four o'clock flower' or 'marvel of Peru' or '*Mirabilis jalapa*' to be awarded the mark.

Inheriting Alleles

Your notes

Phenotypic Plasticity

- Phenotypic plasticity is the idea that although **genotype remains fixed** throughout an organism's lifetime, the way that the **phenotype is expressed** can **vary** during this time
- An organism's internal or external **environment can influence gene expression** patterns, and therefore phenotype
- The levels of regulatory proteins or transcription factors can be affected **in response to environmental stimuli** such as light, and chemicals including **drugs** and **hormones**
- For example, enzymes are activated in response to ultraviolet radiation and increase the **expression and production of melanin,** leading to skin pigmentation
- **Temperature** can also influence gene expression as demonstrated by organisms
  - The Himalayan rabbit (*Oryctolagus cuniculus L.*) possesses a gene for the development of pigmentation in its fur
    - The gene is inactive above 35°C but active between 15°C and 25°C
    - In the parts of the body that are cooler such as ears, feet and nose the gene becomes active making these areas black

Inheriting Recessive Alleles: Phenylketonuria

Your notes

- **Phenylketonuria** (PKU) is an inherited condition caused by a **recessive allele** on an autosome
- It is a condition that can lead to symptoms such as mental disorders and seizures
- It is caused by a build-up of the **amino acid phenylalanine** in the body
  - Phenylalanine comes from broken down protein from diet and our cells
  - The enzyme phenylalanine hydroxylase breaks down phenylalanine
  - This enzyme is coded for by the PAH gene
  - PKU is caused by a **mutation** to the PAH gene that results in a **non-functional enzyme** so that the phenylalanine does not get broken down
- In the UK around 1 in 10,000 people are born with PKU

- In order for a child to have PKU, they must first inherit **two recessive alleles** from each of their parents
- Because it is caused by a recessive allele it means that two non-PKU sufferers could have a child with PKU if **both** parents are **heterozygous carriers** of the mutated PAH gene
- An example genetic cross is shown below:

A genetic cross between two PKU carrier parents diagram

Your notes

	Father	Mother
Parental Phenotypes	Non-PKU	Non-PKU
Parental Genotypes	Pp	Pp
Gametes	(P)(p)	(P)(p)

		Non-PKU Mother	
		(P)	(p)
Non-PKU Father	(P)	PP	Pp
	(p)	Pp	pp

Diagram showing the parental phenotypes, genotypes, gametes and a Punnett square predicting the possible genotypes of their offspring. Both parents are PKU carriers and their offspring have a 25% chance of inheriting the disorder.

- The genetic cross shown on the **Punnett square** above demonstrates that the offspring of the PKU carrier parents have a **75% chance of not having PKU** and a **25% chance of inheriting 2 PKU alleles** and therefore having the condition
- This pattern of inheritance is the same with any autosomal **recessive condition**, for example cystic fibrosis
- Every baby born in the UK and in many other countries around the world are **tested** for several genetic conditions including PKU
  - The babies have a small prick of blood taken from the sole of their foot a few days after being born in order to be screened for the condition

Exam Tip

It should be the case that in most exams letters will be chosen for genetic crosses that have very different upper and lowercase appearances. If you are ever asked to use a letter in an exam that has a similar upper and lowercase appearance, such as P and p, make sure to overly exaggerate the difference to ensure there is no ambiguity during marking.

Your notes

Single Nucleotide Polymorphisms & Multiple Alleles

- Many genes have **more than two alleles**
- However, a **diploid individual** will still **only inherit two of the possible alleles**
- Alleles differ from each other by **one or only a few bases**
- Even a very **small change in base sequence** can bring about a **large effect in gene function**, with a large knock-on effect on the phenotype
- Even though different alleles of a gene have slightly **different base sequences,** they still occupy the **same** locus on the chromosome
- Since the Human Genome Project, **sophisticated techniques** can analyse different alleles
- The exact positions where bases differ between alleles are called **SNPs** or **snips** (Single Nucleotide Polymorphisms)
  - An allele **can have several SNPs** but still only **differ by a few bases** from its other allele

Multiple Alleles: ABO Blood Groups

Your notes

- Inheritance of blood group is an example of **co-dominance** with **multiple alleles**
- This is of critical importance when deciding to give **blood transfusions** following injury or illness
- Use of the wrong blood group can cause an immune response that coagulates (solidifies) blood, leading to clots and serious illness/death
- There are **three alleles** of the gene controlling a person's blood group instead of the usual two
  - I represents the **gene**
  - **Superscripts** A and B represent the **codominant** alleles, I^A for example
  - Lowercase **i** with no superscript represents the **recessive** allele
- I^A results in the production of **antigen A** on the surface of red blood cells
- I^B results in the production of **antigen B** on the surface of red blood cells
- i results in **no antigens** being produced on the surface of red blood cells
- These three possible alleles can give us the following genotypes and phenotypes

Blood Genotype & Phenotype Table

Genotype	Phenotype
I^AI^A or I^Ai	A
I^BI^B or I^Bi	B
I^AI^B	AB
ii	O

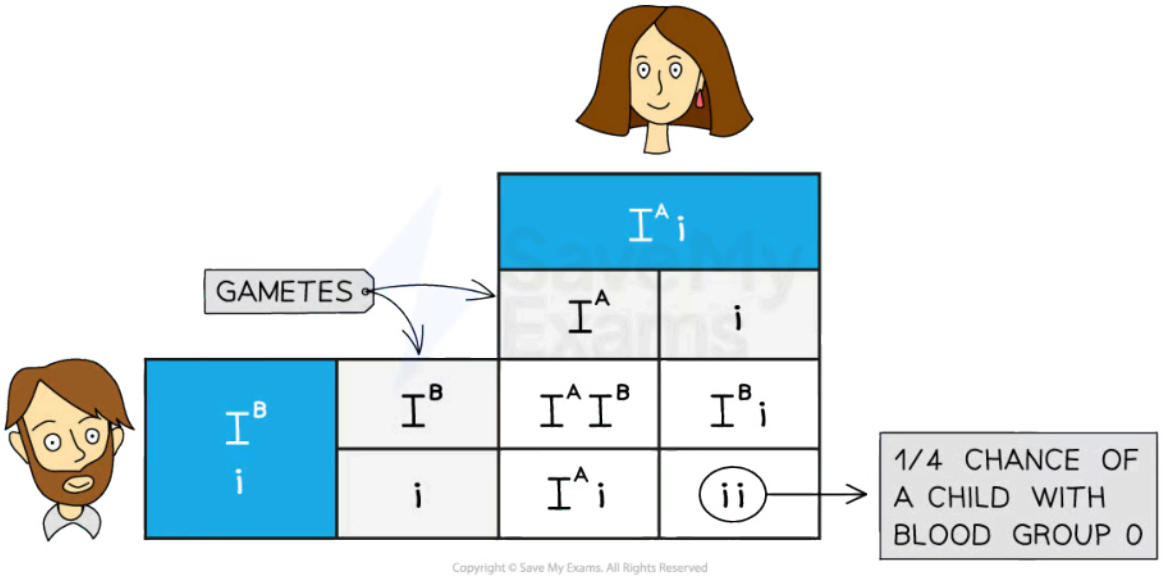
- We can use genetic diagrams to predict the outcome of crosses that involve the codominant alleles controlling blood groups

Worked example

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

Punnett square of the inheritance of blood group

Your notes



Punnett square showing the inheritance of blood group with two heterozygous parents, type A and type B

Sex Determination

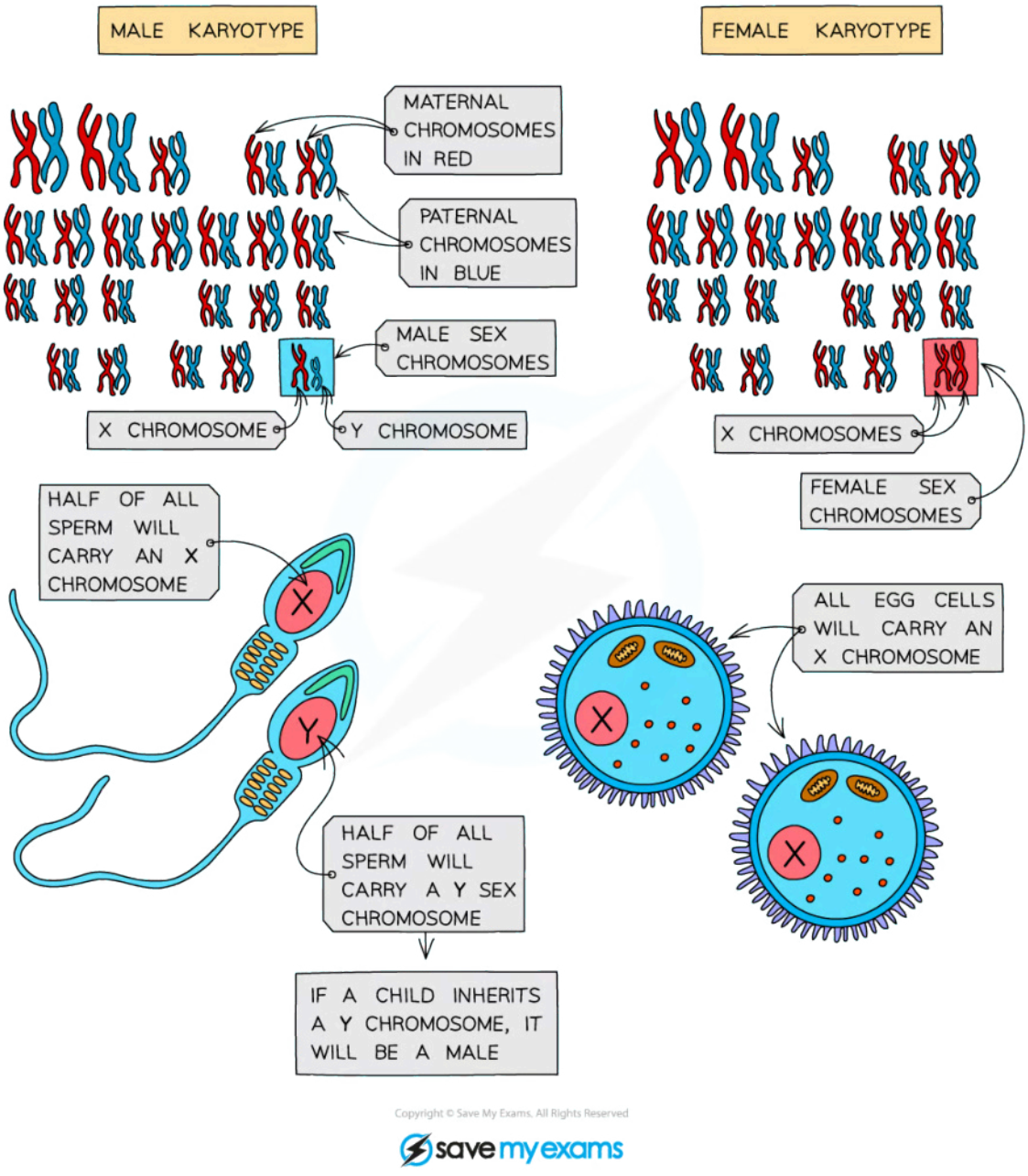

Your notes

Sex Determination in Humans

- 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 (pair 23 in humans) **XX**
- **Males** have the sex chromosomes (pair 23 in humans) **XY**
  - Note that the rule XX for females and XY for males applies to mammals, but not to all species
- All other chromosomes (pairs 1 - 22 in humans) are autosomes and have **no influence on determining the sex** of offspring
- Because only a father can pass on a Y chromosome, he is **responsible for determining the sex of the child**
  - Due to **meiosis**, half of his sperm cells will carry his X chromosome, half his Y chromosome
  - The chromosome carried by **the sperm that fertilises the egg** will determine the sex of the child
  - His daughters receive a copy of his X chromosome
  - His sons receive a copy of his Y chromosome

Sex determination in humans diagram

Your notes

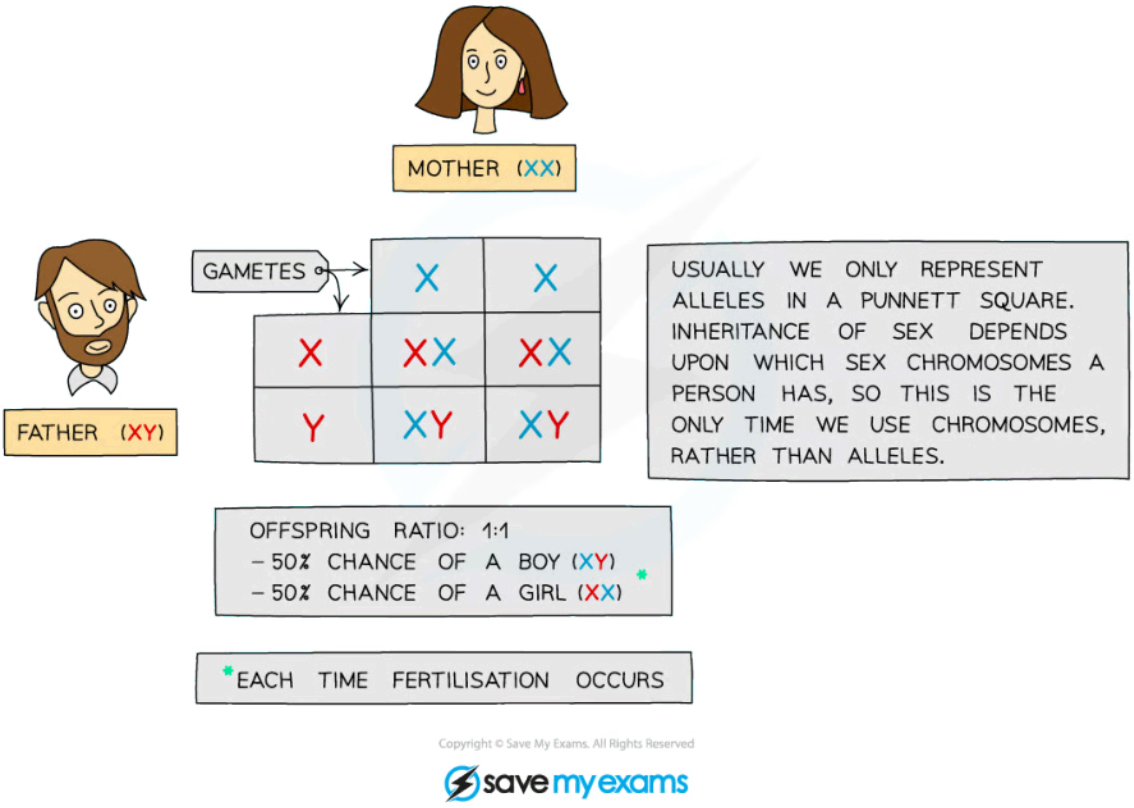


Sperm cells determine the sex of offspring

- 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

Sex determination Punnett square



Punnett square showing the inheritance of sex due to the combination of the X and Y chromosomes from each of the gametes

Genes carried by X and Y chromosomes

- The **X chromosome is larger than the Y**, and has its centromere more central than on the Y chromosome
  - Fewer genes are coded for on the Y chromosome as a result
    - The **X carries around 16 × more genes than the Y** chromosome
  - Non-sex phenotypic traits, including **certain blood clotting factors,** are coded for on the X chromosome but not on the Y
- The Y chromosome carries genes that **code for male characteristics**
- One of these genes is the **SRY gene** which is involved in
  - Development of testes in male embryos
  - Production of testosterone
- Females don't receive these genes, so instead, **ovaries develop** and **female sex hormones** are expressed

Sex Linked Disorders: Haemophilia


Your notes

- Some genetic diseases in humans are sex-linked
- Inheritance of these diseases is different in males and females
  - Sex-linked genes are only **present on one sex chromosome and not the other**
  - This means the sex of an individual affects what alleles they pass on to their offspring through their gametes
- If the **gene is on the X chromosome**, males (XY) will only have one copy of the gene, whereas females (XX) will have two
- There are **three phenotypes for females:**
  - normal
  - carrier
  - has the disease,
- **Males have only two phenotypes**
  - norma
  - has the disease

- **Haemophilia** is a well known sex-linked disease
- There is a gene **found on the X chromosome** that codes for a protein called factor VIII. Factor VIII is needed to make blood clot
- There are two alleles for factor VIII
  - The **dominant F** allele which codes for **normal factor VIII**
  - The **recessive f** allele which results in a **lack of factor VIII**, meaning a person has haemophilia
- When a person possesses only the recessive allele **f**, they don't produce factor VIII and their blood can't clot normally
- If males have an abnormal allele, f, they will **have the condition** as they have only one copy of the gene
- Females can be heterozygous for the faulty gene and not suffer from the condition but act as a **carrier**
- This means that haemophilia is a potentially fatal genetic disease which **affects males more than females**

Exam Tip

The expected notation when writing about sex linked alleles is to use upper case 'X' and 'Y' for the chromosome, next to superscript letters to represent the allele. For example

- X^fX^f Homozygous female who has haemophilia or X^FX^f Heterozygous female who is a carrier
- X^fY Male who has haemophilia

Your notes

Worked example

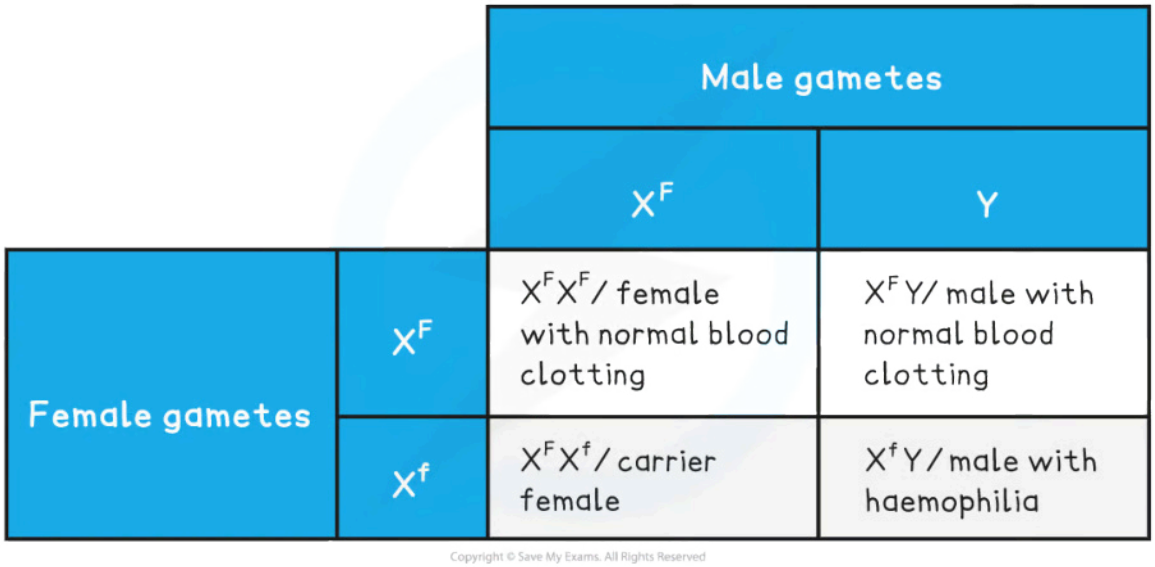
The genetic diagram below shows how two parents with normal factor VIII can have offspring with haemophilia

Parental phenotypes: carrier female x normal male

Parental genotypes: X^FX^f X^FY

Parental gametes: X^F or X^f X^F or Y

Monohybrid Punnett Square with Sex-linkage Table



Predicted ratio of phenotypes in offspring

1 female with normal blood clotting : 1 carrier female : 1 male with haemophilia : 1 male with normal blood clotting

Predicted ratio of genotypes in offspring: 1 X^FX^F : 1 X^FX^f : 1 X^FY : 1 X^fY

Exam Tip

Make sure to include all of your working out when constructing genetic diagrams. It is not enough just to complete a Punnett grid, you need to show that you have thought about the **possible gametes** that can be produced by each parent. Also, remember to state the **phenotype** as well as the genotype of the offspring that result from the cross. Read the questions carefully when answering sex-linked inheritance questions – is the question asking for a probability for **all** children or is it asking about a specific sex (males or females).

Your notes

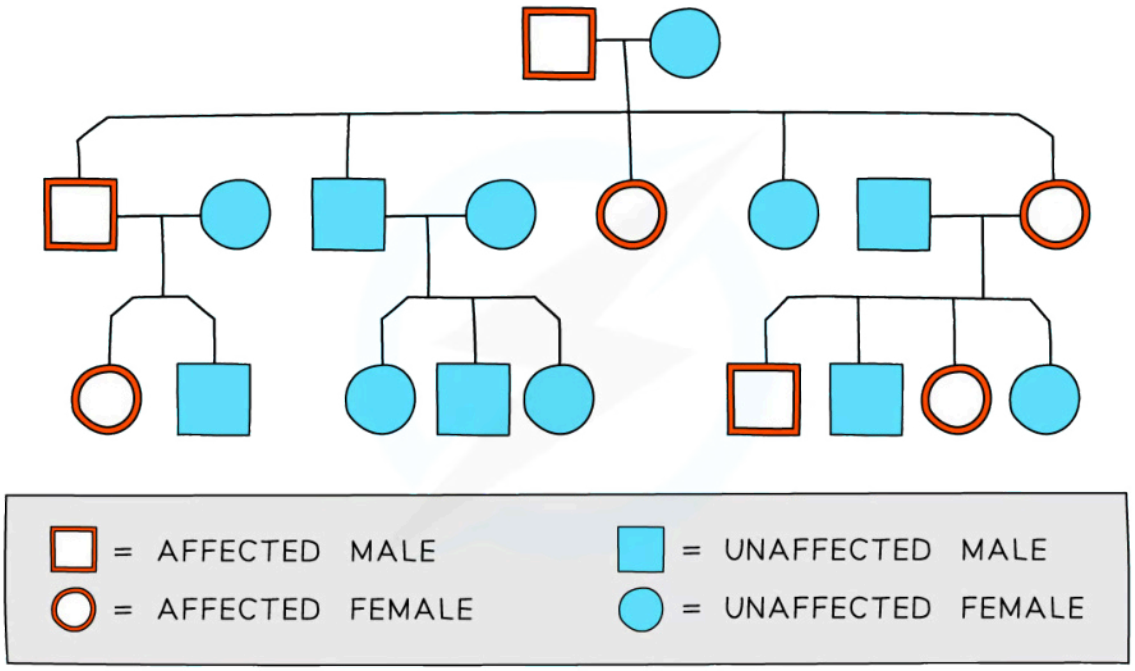
Pedigree Charts

Your notes

Pedigree Charts

- 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**

Pedigree chart diagram



A family pedigree chart

- **Males** are indicated by the **square shape** and **females** are represented by **circles**
- In this diagram, **affected** individuals are **red** and **unaffected** are **blue**
  - **Shading or cross-hatching** may also be used to show affected individuals
- **Horizontal lines** between males and females show that they have **produced children** (which are linked underneath each couple)
- **Roman numerals** may be used to indicate **generations**
- For each generation the eldest child is on the left and **each individual** is **numbered**
- The family pedigree above shows:
  - Both males and females are affected
  - Every generation has affected individuals

- The eldest son (in the second generation) is affected
- 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

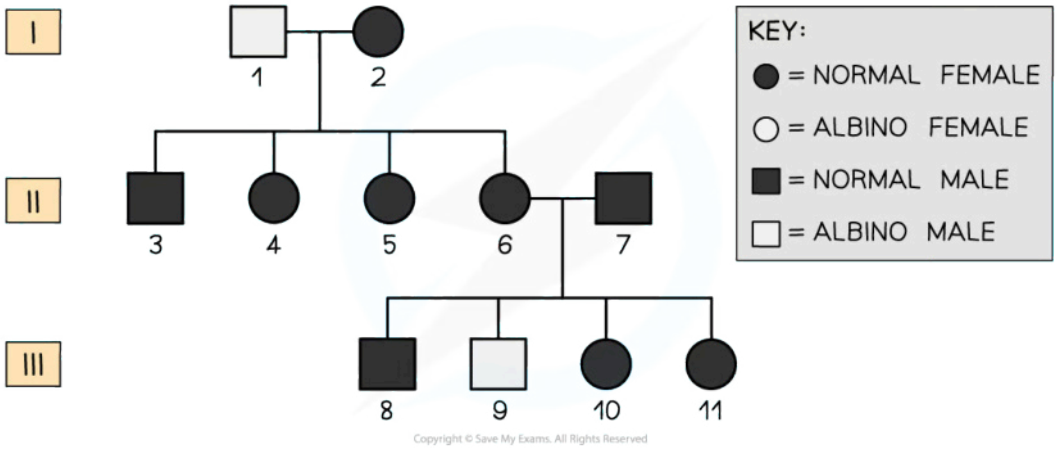
- The study of pedigree charts provides an opportunity to appreciate why marriage between close relatives is **prohibited** in many countries
  - Reproducing with close relatives increases the chance that both individuals possess **harmful recessive alleles** that can be passed onto the offspring
  - This causes the offspring to have a much **higher chance** of inheriting **genetic diseases**

Your notes

Worked example

Worked example: Pedigree charts

- Below is a pedigree chart which traces the inheritance of **albinism** across several generations. Albinism affects the production of the **pigment melanin** leading to lighter hair, skin and eyes.



- Using the pedigree chart, deduce and explain the following:
  1. What type of allele causes albinism?
  2. The genotype of individuals named **9** and **7**
  3. The possible genotypes of **10** and **11**

1. Albinism is caused by a **recessive allele**
   - **Explanation**: We can tell this from the pedigree chart because expression of the disease skips generation II. Also, person number 9 is an affected individual despite his parents (6 and 7) being unaffected. 6 and 7 must both be carriers of the recessive allele and 9 has inherited one recessive allele from each parent.
   - It is unlikely to be a **sex-linked disease as both females and males** have the condition
2. The genotype of person 9 must be **homozygous recessive** (aa) and the genotype of 7 must be **heterozygous** (Aa)
   - **Explanation**: 9 is an affected individual with albinism (which is determined by the recessive allele). 7 must be heterozygous in order for him to pass on the recessive allele to person 9
3. The possible genotypes of 10 and 11 are **heterozygous** (Aa) or **homozygous dominant** (AA)

- **Explanation**: This is because they are unaffected individuals so must possess at least one dominant allele (A), however, it is possible that they each inherited a dominant allele from each parent


Your notes

Exam Tip

When answering questions about pedigree charts for genetic diseases, it is always useful to remember which phenotype is caused by the recessive allele. You can write these genotypes onto your chart and it will give you a good starting point for working out the possible genotypes of the rest of the individuals in the chart.

NOS: Scientists draw general conclusions by inductive reasoning

- **Inductive reasoning** is the idea of making **generalised conclusions** based on **specific evidence** taken from a small sample
  - For example, we could observe a sample of evidence from a pedigree chart and if that observation **deviated from what we would expect** we **could** surmise that the condition could be sex-linked
- **Deductive reasoning** is making **specific deductions** about something unknown based on **known evidence**
  - For example, if two non-affected parents have a child that is affected by a genetic condition we can deduce that the condition is caused by a recessive allele, and that the parents are both carriers of the allele

Continuous Variation: Skills

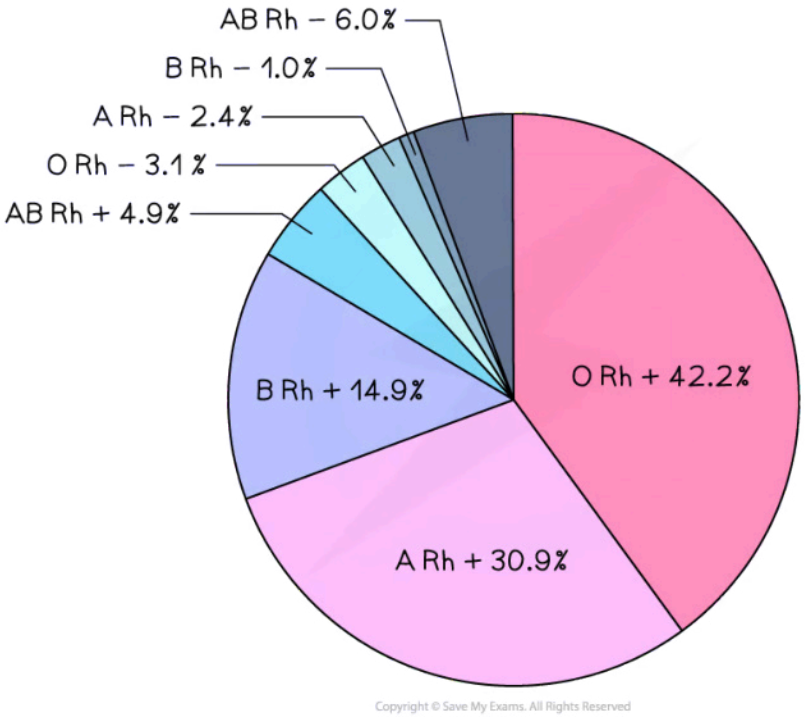
Your notes

Continuous Variation

Variation can be discrete or continuous

- **Discrete variation** is variation that falls 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:

Pie chart showing global blood group distribution



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

(data varies regionally with ethnicity)

Continuous Variation

- **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

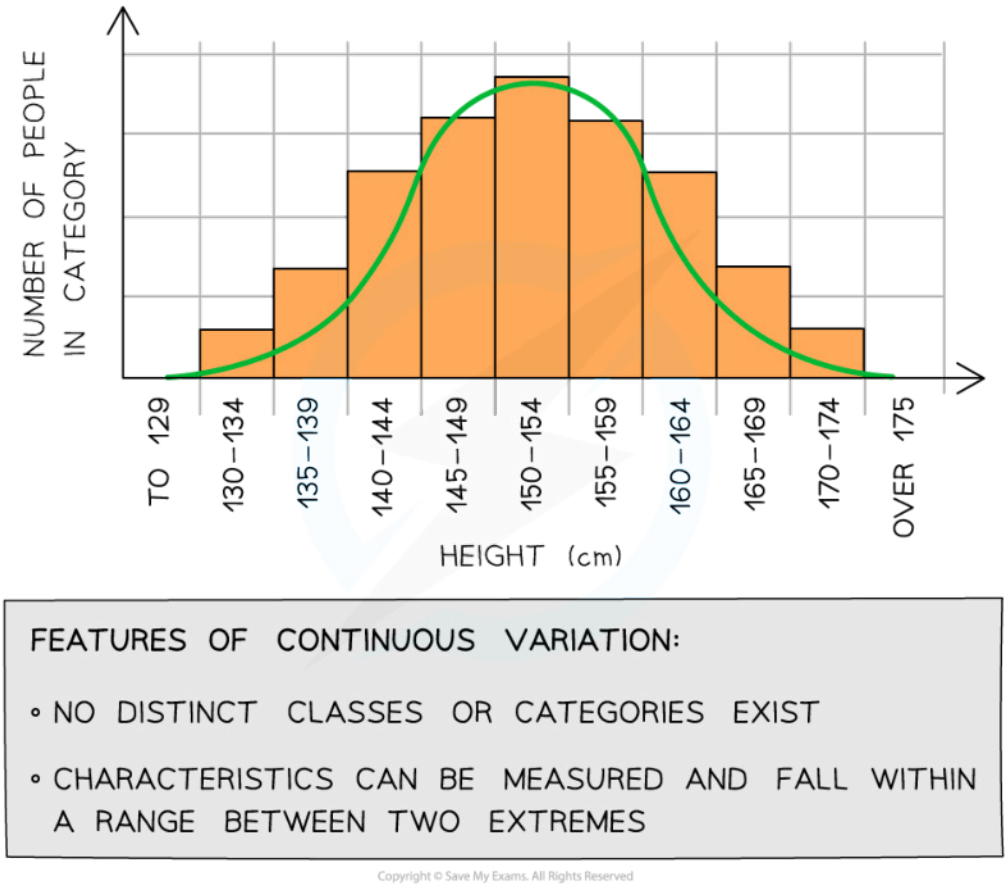
- Hormone production
- ...As well as environmental factors like diet, exercise, prenatal nutrition, lifestyle etc

- 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)
  - Environmental factors can also affect birth mass, e.g. 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

Normal distribution curve

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**
- An example of a **continuous polygenic trait** is **skin colour**
- Skin colour is **determined by several genes** that cause the production of a protein called **melanin**
  - The more melanin is produced, the darker the skin pigmentation becomes
- Skin colour is also influenced by **environmental factors** such as UV exposure, which can cause the skin colour to become darker

Comparison of Continuous and Discontinuous Variation Table

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


Your notes

Box & Whisker Plots


Your notes

What are box plots and when should they be used?

- **Box plots** are also known as **box-and-whisker diagrams**
- They are used when we are interested in splitting data up into **quartiles**
- Using quartiles and drawing a box plot allows us to see what is happening at the **low, middle and high points and consider any possible extreme values**

How to draw a box plot

- You need to know **five values** to draw a box plot
  - Lowest data value
  - First quartile
  - Median
  - Third quartile
  - Highest data value
- Usually on **graph paper**, box plots are drawn accurately with the five points marked by **short vertical lines**
  - The middle three values then form a box with the **median line inside**
    - The median will not necessarily be in the middle of the box
  - The box represents the **interquartile range** (middle 50% of the data)
  - The **lowest** data value and **highest** data value are joined to the box by **horizontal lines**
    - These are often called **whiskers**
    - They represent the **lowest 25%** of the data and the **highest 25%** of the data
- You may be given a box plot
  - From which you can read off the five values
  - Calculate other statistics like the **range** and **interquartile range** (IQR)

Your notes

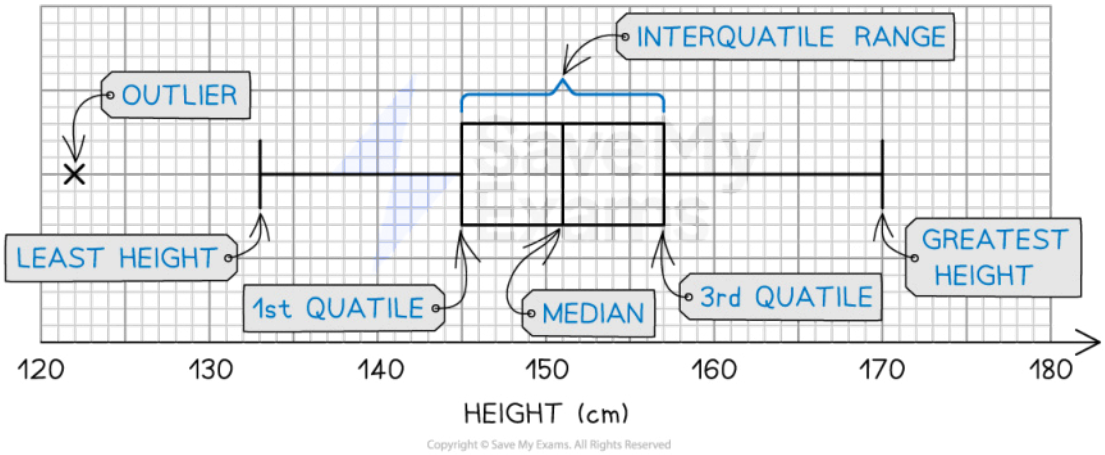
Worked example

The table gives some information about the heights of 80 girls:

	Height (cm)
Least height	133
Greatest height	170
First quartile	145
Third quartile	157
Median	151
Outlier	122

A box plot of this data is as follows:

Box and whisker plot showing height data from a population of girls



The width of the box shows the interquartile range and the overall range is shown by the length of the whiskers

Outliers

- **Outliers** are data points that exist at the **extremes** above and below the rest of the data
- In order for a data point to be categorised as an outlier it needs to be more than **1.5 × the interquartile range** above the third quartile or below the first quartile
- As shown in the worked example above, the outliers are **plotted separately** to the whiskers of the plot


Your notes