



Inheritance

Revision notes · IB Biology D3.2: genetic crosses, dominance, PKU, ABO blood groups, sex linkage, pedigrees and box plots.

For IB Diploma Biology SL undefined. **HL only** is additional higher level content, for HL students only.

Inheritance Spec D3.2

Haploid gametes, diploid zygotes and genetic crosses in plants Spec D3.2.1, D3.2.2

Each parent produces **haploid** gametes by meiosis, carrying **one copy of each gene**. At fertilization two gametes fuse to give a **diploid** zygote with **two copies of each autosomal gene**, one from each parent. This pattern of inheritance is common to all eukaryotes with a sexual life cycle.

Carrying out a cross in flowering plants

Pollen contains the male gametes and the female gametes are in the ovules inside the ovary, so **pollination is needed to carry out a cross**. Plants such as peas produce male and female gametes in the same flower, so they normally **self-pollinate** and self-fertilize.

1. Choose two pure-breeding (homozygous) parents with contrasting traits: the **P generation**.
2. Remove the immature anthers from flowers of one parent so it cannot self-pollinate.
3. Transfer pollen from the other parent to its stigmas with a small brush, then cover the flowers with a bag to stop other pollen arriving.
4. Collect the seeds and grow them: these plants are the **F1 generation**. Record their phenotypes.
5. Allow the F1 plants to self-pollinate (or cross them with each other). The offspring are the F2 generation. Count each phenotype.
6. Use a **Punnett grid** to predict the expected ratios and compare them with the counts.

Genetic crosses like these are widely used to breed new varieties of crop and ornamental plants, combining useful traits such as yield, disease resistance or flower colour.

Exam tip: Always show a full genetic diagram: parental phenotypes and genotypes, gametes, a Punnett grid, then offspring genotypes, phenotypes and ratio.

Genotype, phenotype, dominance and phenotypic plasticity Spec D3.2.3, D3.2.4, D3.2.5, D3.2.6

Gene	A heritable factor that occupies a specific position (locus) on a chromosome and influences a characteristic.
Allele	One of the alternative forms of a gene, differing in base sequence by one or a few bases.
Genotype	The combination of alleles of a gene (or genes) inherited by an organism.
Homozygous	Having two identical alleles of a gene, for example AA or aa.
Heterozygous	Having two different alleles of a gene, for example Aa.
Phenotype	The observable traits of an organism, resulting from its genotype and environmental factors.

Causes of phenotype in humans	
Cause	Examples
Genotype only	ABO blood group; inherited conditions such as haemophilia
Environment only	Scars, tattoos, the language a person speaks
Interaction of genotype and environment	Height and body mass (genes plus nutrition); skin colour (genes plus exposure to sunlight)

Dominant and recessive alleles

A **dominant allele** usually codes for a functional protein, often an enzyme. One copy produces enough of it for the trait to appear, so **homozygous dominant (AA) and heterozygous (Aa) individuals have the same phenotype**. A **recessive allele** often codes for a non-functional protein, or none, so its effect only shows when there is no dominant allele (aa).

Phenotypic plasticity

Phenotypic plasticity is the capacity to develop traits suited to the environment an organism experiences, by **varying patterns of gene expression**. The genotype does not change, and the changes may be **reversible** during the individual's lifetime.

- Human skin tans in strong sunlight as melanin production increases, then fades in winter.
- Skeletal muscles grow larger with regular training and shrink when training stops.
- Water crowfoot (*Ranunculus aquatilis*) grows finely divided leaves under water and broad leaves above it, from the same genotype.

Common mistake: Phenotypic plasticity is not mutation or evolution. The alleles stay the same; only which genes are expressed, and how much, changes.

Phenylketonuria, SNPs, multiple alleles and ABO blood groups Spec D3.2.7, D3.2.8, D3.2.9

Phenylketonuria

Phenylketonuria (PKU) is a recessive genetic condition caused by a mutation in an **autosomal gene** that codes for the enzyme phenylalanine hydroxylase, needed to **convert phenylalanine to tyrosine**.

- Only people with two recessive alleles (pp) have PKU. Heterozygotes (Pp) make enough enzyme from their one functional allele, so they are unaffected carriers.

- Without the enzyme, phenylalanine from dietary protein builds up in the blood and damages the developing brain, causing intellectual disability. Less tyrosine is made, so less melanin.
- Babies are screened soon after birth with a blood test. Affected children follow a diet low in phenylalanine, which prevents most harm: an example of environment affecting phenotype.

Single-nucleotide polymorphisms and multiple alleles

A **single-nucleotide polymorphism (SNP)** is a position in a gene where the base differs between individuals. Each new SNP arising by mutation can create a new allele, so **any number of alleles** of a gene can exist in the **gene pool** (all the alleles in an interbreeding population). A diploid individual, though, only inherits **two** alleles of each autosomal gene.

ABO blood groups

The ABO gene has three common alleles. **I^A** and **I^B** code for enzymes that add different sugars to a glycoprotein antigen on red blood cells; **i** codes for a non-functional enzyme. I^A and I^B are both dominant to i, and codominant with each other.

ABO genotypes and phenotypes		
Blood group	Genotypes	Antigen on red blood cells
A	I ^A I ^A or I ^A i	A
B	I ^B I ^B or I ^B i	B
AB	I ^A I ^B	A and B
O	ii	Neither

An ABO cross

A father of blood group A (I^Ai) and a mother of blood group B (I^Bi) have a child. Determine the possible blood groups of their children and the probability of each.

1. Father's gametes: I^A or i. Mother's gametes: I^B or i.
2. Punnett grid: I^AI^B, I^Ai, I^Bi, ii.
3. Phenotypes: AB, A, B, O, one of each.

Answer: Each child has a 0.25 (25%) chance of each blood group: A, B, AB or O.

Exam tip: Use exactly the notation I^A, I^B and i for ABO alleles. Writing A, B and O as alleles usually loses the mark.

Incomplete dominance and codominance Spec D3.2.10

Patterns of inheritance at the phenotypic level		
Pattern	Heterozygote phenotype	Example
Complete dominance	Same as homozygous dominant	Pea plants: Tt is tall, like TT
Incomplete dominance	Intermediate between the two homozygotes	Four o'clock flower (<i>Mirabilis jalapa</i>): red × white gives pink
Codominance	Dual phenotype: both alleles expressed	Blood group AB (I ^A I ^B): both A and B antigens present

Incomplete dominance in the four o'clock flower

A pure-breeding red *Mirabilis jalapa* ($C^R C^R$) is crossed with a pure-breeding white plant ($C^W C^W$). The F1 are self-pollinated. Deduce the F1 and F2 phenotypes and ratio.

1. P gametes: C^R and C^W , so all F1 are $C^R C^W$: pink.
2. F1 $\times$ F1: each produces C^R and C^W gametes in equal numbers.
3. Punnett grid: $C^R C^R$, $C^R C^W$, $C^W C^R$, $C^W C^W$.

Answer: F1 all pink. F2 1 red : 2 pink : 1 white; the genotype ratio and phenotype ratio are the same.

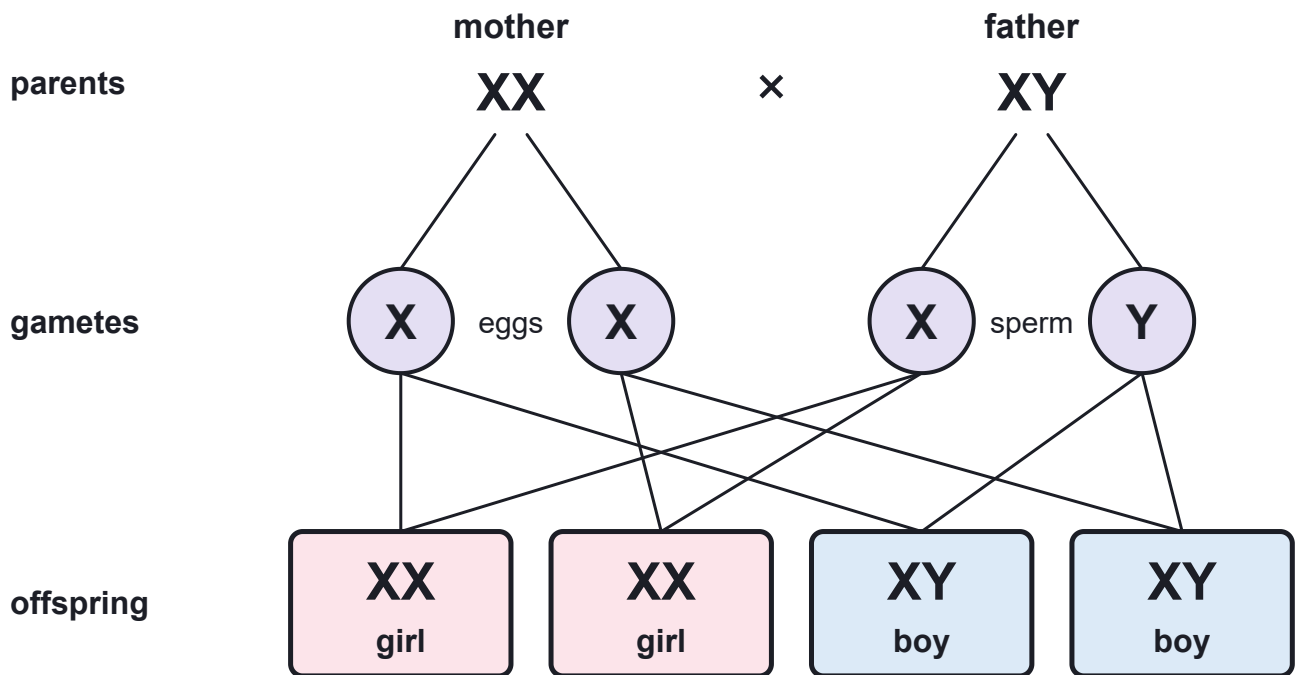
In the pink heterozygote, the single C^R allele produces only enough red pigment to make the petals pink. In blood group AB, each allele makes its own enzyme, so both antigens appear side by side.

Exam tip: A 1:2:1 phenotype ratio in an F2 is a sign of incomplete dominance or codominance, because the heterozygotes can be told apart. Either the common or the scientific name of the organism is acceptable.

Sex determination, sex linkage and haemophilia Spec D3.2.11, D3.2.12

Human body cells have 22 pairs of autosomes and one pair of **sex chromosomes**: **XX** in female-typical and **XY** in male-typical individuals. Every egg carries an X. Half the sperm carry an X and half a Y, so **the sex chromosome in the sperm determines** the development of the zygote.

- A gene on the Y chromosome (SRY) causes the embryonic gonads to develop into testes. Testosterone from the testes then leads to male-typical characteristics. Without it, ovaries and female-typical characteristics develop.
- The X chromosome is much larger and carries **far more genes** than the Y chromosome, most of them nothing to do with sex.
- Genes carried on the X chromosome are **sex-linked**. A male has only one X, so whichever allele he has on it is expressed, even if recessive.



2 girls : 2 boys, a ratio of 1 : 1

50% chance of a girl and 50% chance of a boy

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Inheritance of sex: the sperm carries X or Y, so each child has a **50% chance** of XX (female-typical) or XY (male-typical).

Haemophilia

In **haemophilia** the blood clots very slowly because a clotting protein (factor VIII in the commonest type) is missing. The gene is on the X chromosome. The normal allele is **X^H** and the recessive haemophilia allele **X^h**.

Haemophilia genotypes

Genotype	Phenotype
X^HX^H	Unaffected female
X^HX^h	Unaffected carrier female
X^hX^h	Affected female (rare)
X^HY	Unaffected male
X^hY	Affected male

Carrier mother and unaffected father

A carrier woman (X^HX^h) and an unaffected man (X^HY) have children. Deduce the probabilities of the possible outcomes.

1. Mother's gametes: X^H or X^h. Father's gametes: X^H or Y.
2. Punnett grid: X^HX^H, X^hX^H, X^HY, X^hY.
3. Daughters: half unaffected, half carriers. Sons: half unaffected, half affected.

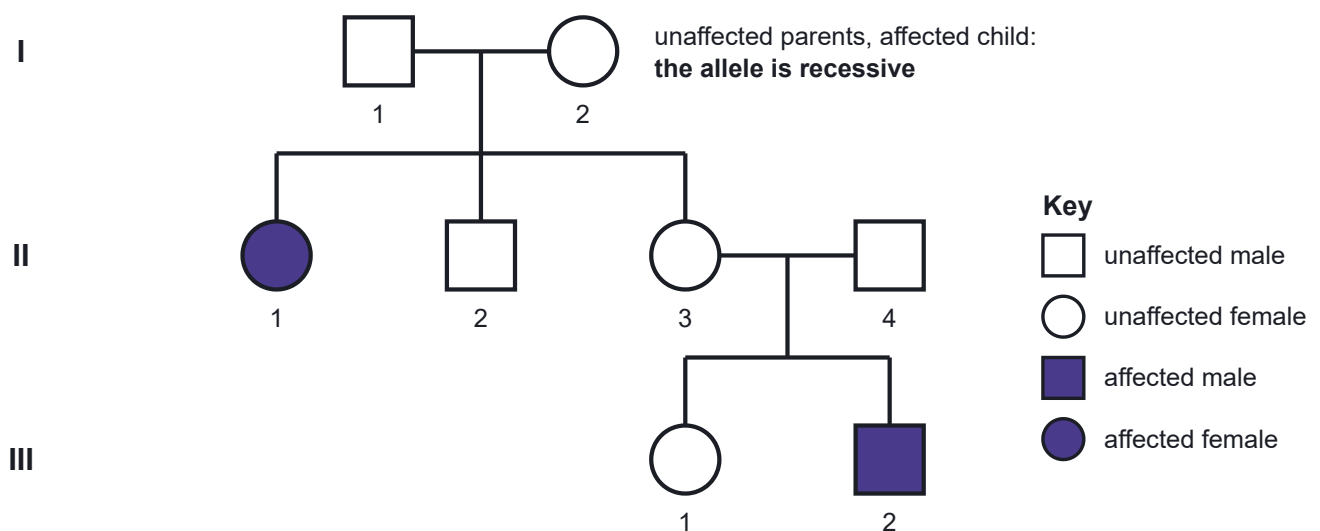
Answer: Probability a child is an affected son = 0.25; probability that a son is affected = 0.5; no daughters affected.

Common mistake: A father cannot pass an X-linked allele to his sons, because sons receive his Y. Always write sex-linked alleles as superscripts on an uppercase X.

Pedigree charts Spec D3.2.13

In a **pedigree chart**, squares are males and circles are females, shaded if affected. A horizontal line joins parents, and a vertical line leads down to their children. Generations are numbered with Roman numerals (I, II, III) and individuals with numbers.

Clues to the pattern of inheritance	
Pattern	Clues in the pedigree
Autosomal recessive	Two unaffected parents have an affected child, so both parents must be carriers; males and females affected equally
Autosomal dominant	Every affected individual has at least one affected parent; two affected parents can have an unaffected child
Sex-linked (X-linked) recessive	Mostly males affected, usually with unaffected carrier mothers; an affected father never passes it to his sons



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A pedigree for an **autosomal recessive** condition. Unaffected parents I-1 and I-2 have an affected child, so both must be carriers.

Inductive and deductive reasoning

Working out the pattern from part of a pedigree is **inductive reasoning**: a general conclusion (a theory) from some but not all cases. Using that theory to work out the genotypes of specific individuals is **deductive reasoning**.

Marriage between close relatives

Close relatives share many alleles inherited from common ancestors. If both partners carry the **same rare recessive allele**, each child has a 0.25 chance of being homozygous recessive and affected. This is the genetic basis for the prohibition of marriage between close relatives in many societies.

Exam tip: When deducing a pattern, quote the individuals: for example, 'II-1 is affected but I-1 and I-2 are not, so the allele is recessive'.

Continuous variation and box-and-whisker plots Spec D3.2.14, D3.2.15

Polygenic inheritance is the control of one trait by **many genes**, each with a small additive effect. With many genes the number of possible genotypes is large, so phenotypes form a continuous range, usually close to a normal distribution. Environmental factors add further variation.

- **Skin colour** in humans is polygenic: several genes affect the amount and type of melanin. Environment also matters, because exposure to sunlight increases melanin production.
- A **continuous variable**, such as skin colour or height, can take any value in a range.
- A **discrete variable**, such as ABO blood group, falls into distinct categories with no intermediates.

Mean	The sum of all values divided by the number of values.
Median	The middle value when the data are in order.
Mode	The most frequent value (or class).

Box-and-whisker plots

A **box-and-whisker plot** displays six aspects of a data set: **outliers, minimum, first quartile (Q1), median, third quartile (Q3) and maximum**. The box runs from Q1 to Q3 with a line at the median. The whiskers reach the lowest and highest values that are not outliers.

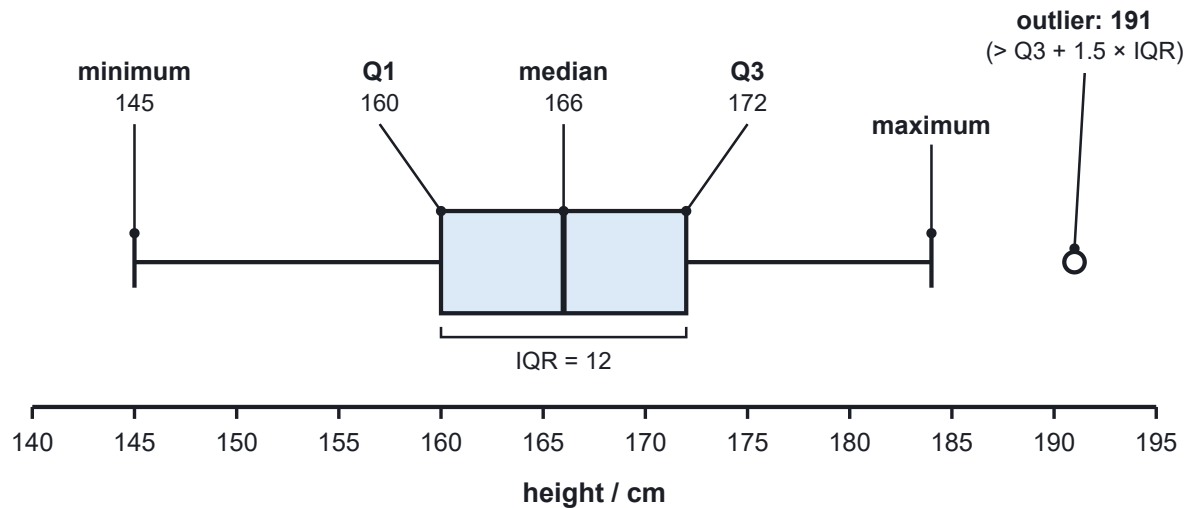
interquartile range (IQR) = $Q3 - Q1$; outlier if $> Q3 + 1.5 \times IQR$ or $< Q1 - 1.5 \times IQR$

Outliers in student height data

Heights of a class of students (cm): $Q1 = 160$, median = 166, $Q3 = 172$. The shortest student is 145 cm and the tallest is 191 cm. Determine whether either is an outlier.

1. $IQR = 172 - 160 = 12$ cm.
2. $1.5 \times IQR = 18$ cm.
3. Upper limit = $172 + 18 = 190$ cm. Lower limit = $160 - 18 = 142$ cm.
4. $191 \text{ cm} > 190 \text{ cm}$, so it is an outlier. $145 \text{ cm} > 142 \text{ cm}$, so it is not.

Answer: 191 cm is an outlier (plotted as a separate point); the upper whisker ends at the tallest non-outlier. 145 cm is the minimum.



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The six aspects of a **box-and-whisker plot**, using the student height data from the worked example. 191 cm is beyond 190 cm ($Q3 + 1.5 \times IQR$), so it is plotted as an outlier.

Maths skill: Box plots let you compare two samples quickly: compare medians for the typical value and IQRs for spread. State the units every time.

Exam tip: Linking question: what are the principles of effective sampling in biological research? A larger random sample gives quartiles and medians that better represent the population.

Quick check

1. Why do AA and Aa individuals have the same phenotype?
2. Give the genotype of a person with blood group O.
3. What phenotype does a heterozygote show in incomplete dominance?
4. State the outlier rule for a box-and-whisker plot.

Quick check answers

Inheritance

1. One dominant allele produces enough functional protein for the trait to appear.
2. ii.
3. An intermediate phenotype, e.g. pink four o'clock flowers.
4. More than $1.5 \times \text{IQR}$ above Q3 or below Q1.