

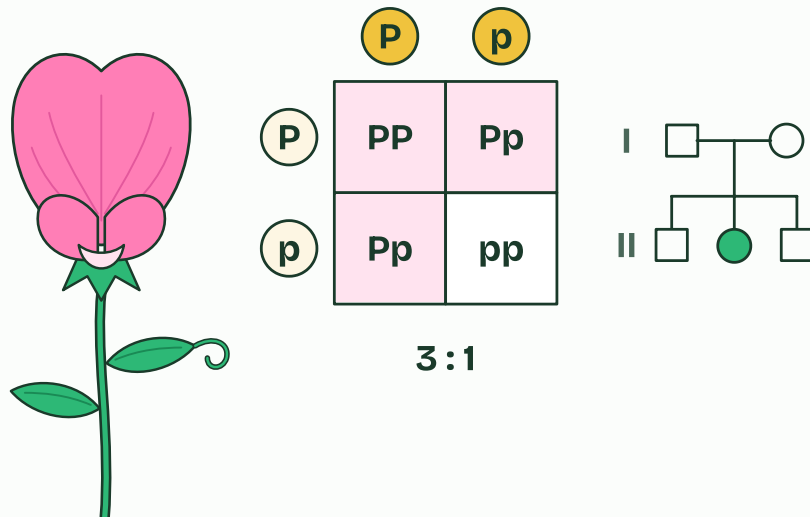
REVISION GUIDE

How traits are *handed* *down*

Inheritance

Half of your genes came from each parent, and which half is a matter of chance. Follow the alleles from the gamete to the grid to the family tree, learn why some traits hide for a generation, and at higher level test whether two genes really travel apart.

The same pattern in a family



Contents

01	Halve, then fuse	4
02	How to make a cross	8
03	Genes, alleles and the traits you see	11
04	Dominant and recessive alleles	13
05	Same genes, different outcomes	16
06	One faulty enzyme: PKU	18
07	Many alleles, not just two	20
08	Neither wins, or both do	22
09	X, Y and sex	24
10	Reading a family tree	26
11	A spread, not a few types	29
12	Showing a spread with a box plot	31
13	Two genes at once HL	34
14	Genes that travel together HL	39
15	Is it chance? The chi-squared test HL	42
	Key terms	45
	Dry lab: Fly crosses: linked or not?	48
	Check yourself	51
	Answers	52

By the end you can

- ☐ Explain how meiosis halves the chromosome number to make haploid gametes, and how fusion gives a diploid zygote D3.2.1
- ☐ Describe how a genetic cross is made in flowering plants, using the terms P generation, F1 generation, F2 generation and Punnett grid D3.2.2
- ☐ Define genotype, and use homozygous, heterozygous, gene and allele correctly D3.2.3
- ☐ Explain how phenotype comes from genotype and the environment, with examples of each D3.2.4
- ☐ Predict the effects of dominant and recessive alleles on phenotype D3.2.5
- ☐ Explain phenotypic plasticity as a change in gene expression, not in the genes themselves D3.2.6
- ☐ Explain phenylketonuria as a disease caused by a recessive allele D3.2.7
- ☐ Describe single-nucleotide polymorphisms and multiple alleles in a gene pool D3.2.8
- ☐ Use the ABO blood groups to show how multiple alleles are inherited D3.2.9
- ☐ Distinguish incomplete dominance from codominance D3.2.10
- ☐ Explain sex determination in humans and the inheritance of genes on the sex chromosomes D3.2.11
- ☐ Use haemophilia to explain a sex-linked genetic disorder D3.2.12
- ☐ Use pedigree charts to deduce patterns of inheritance of genetic disorders D3.2.13
- ☐ Explain continuous variation as the result of polygenic inheritance and the environment D3.2.14
- ☐ Draw and read box-and-whisker plots for a continuous variable such as student height D3.2.15
- ☐ Explain segregation and independent assortment of unlinked genes in meiosis D3.2.16
- ☐ Use Punnett grids to predict genotypic and phenotypic ratios in dihybrid crosses of unlinked autosomal genes D3.2.17
- ☐ Find the loci of human genes and their polypeptide products in a database D3.2.18
- ☐ Explain autosomal gene linkage D3.2.19
- ☐ Identify recombinants in crosses involving two linked or unlinked genes D3.2.20
- ☐ Use a chi-squared test on data from a dihybrid cross D3.2.21

Tick each one when you can do it without the book.

01

D3.2.1 HALVING AND DOUBLING

Halve, then *fuse*

Your body cells hold two copies of every chromosome, one set from each parent. A cell like that is **diploid**. A human body cell has 46 chromosomes, written $2n = 46$. Most of them are **autosomes**, so a diploid cell has two copies of each autosomal gene: one on each chromosome of a pair.

If a child simply received all the chromosomes of both parents, the number would double every generation. Life avoids this by halving first. Special cells divide by **meiosis** to make **gametes**, and each gamete gets one chromosome from every pair. A cell with a single set is **haploid**. In humans, $n = 23$.

Then fertilisation joins two gametes. Their nuclei fuse to make a **zygote**, which is diploid again, with one set from each parent. Halve, then double: that is the loop that carries genes from one generation to the next.

This pattern is common to all eukaryotes with a sexual life cycle. You, a pea plant, a mushroom and a fruit fly all make haploid gametes and fuse them to form a diploid zygote. The details of the life cycle differ from group to group, but the loop is the same.

KEY TERMS

diploid

A cell with two sets of chromosomes, one from each parent ($2n$). It carries two copies of each autosomal gene.

autosomes

Any chromosome that is not a sex chromosome. Autosomes come in matching pairs.

meiosis

A type of cell division that halves the chromosome number. It makes the gametes of an organism that reproduces sexually.

gametes

A sex cell, such as a sperm, an egg or a pollen grain, with one set of chromosomes.

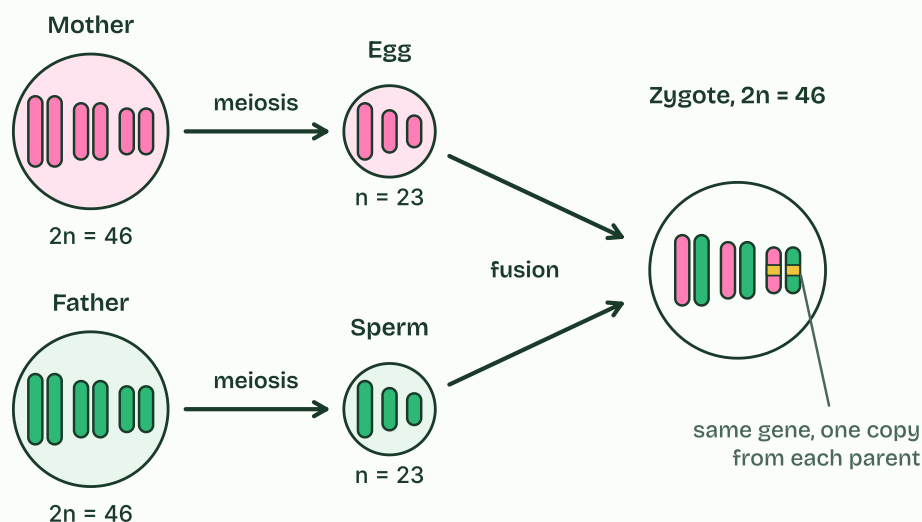
haploid

A cell with one set of chromosomes (n), such as a gamete.

zygote

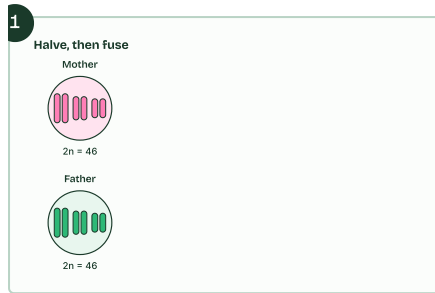
The diploid cell made when two gametes fuse. It divides and grows into a new organism.

Halve, then fuse

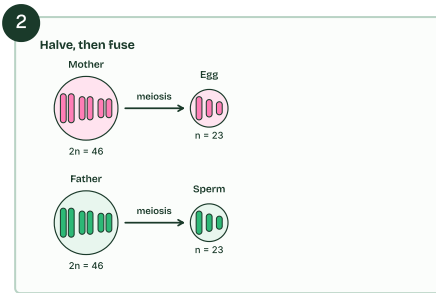


STEP BY STEP

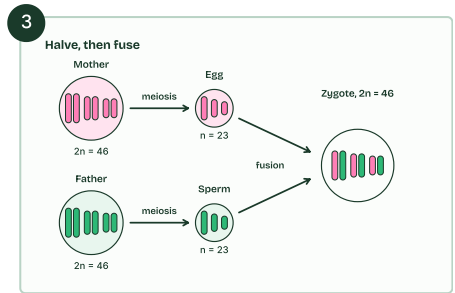
4 STEPS



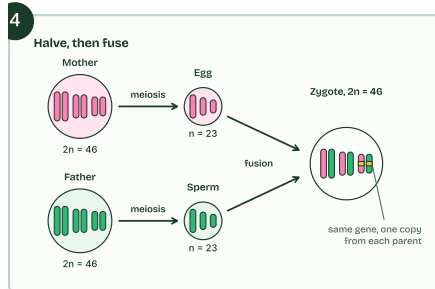
Two parents, each with **diploid** body cells ($2n = 46$). Three of the 23 pairs are drawn.



Meiosis makes gametes with one chromosome from each pair: an egg and a sperm, each **haploid** ($n = 23$).



Fusion of the two gametes makes a zygote. It is diploid again, with one set from each parent.

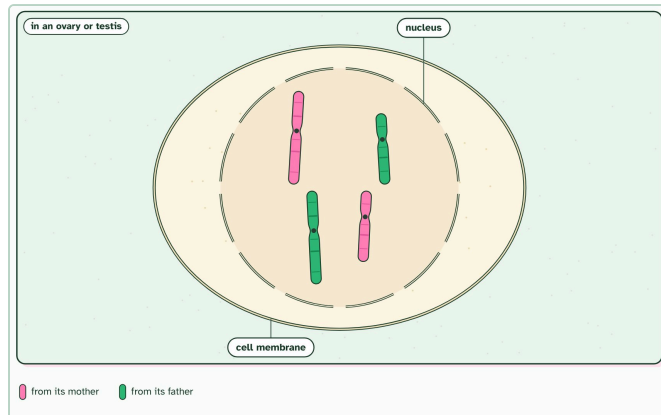


In the zygote, a pair of chromosomes carries the **same gene** twice, one copy from each parent.

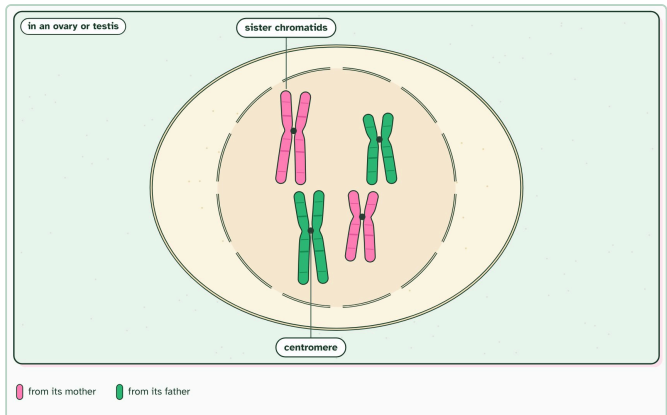
ANIMATION · WATCH IT HAPPEN: HALVING, THEN FUSING

9 MOMENTS

1 DIPLOID CELL, $2N = 4$ 2 $2N = 4$, DNA COPIED

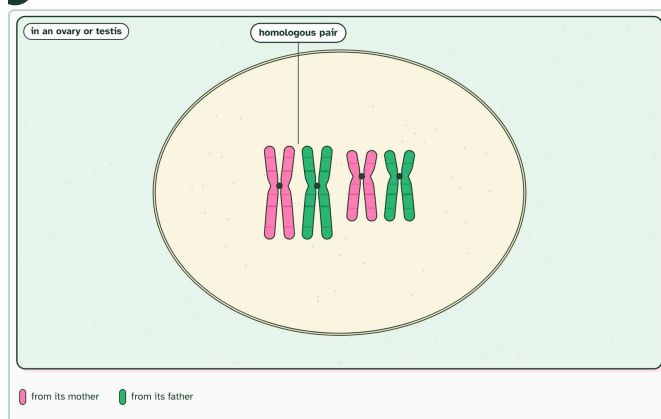


A **diploid** cell of a parent. Its chromosomes are in pairs: one of each pair came from its mother (pink), one from its father (green).

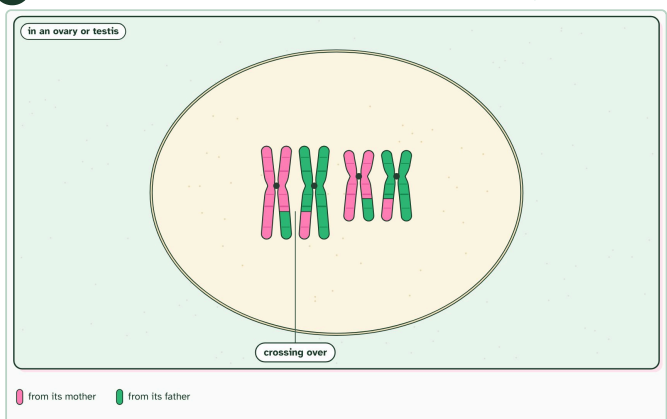


First the DNA is copied: each chromosome becomes two identical **sister chromatids**, joined at a **centromere**.

3 $2N = 4$, DNA COPIED 4 $2N = 4$, DNA COPIED

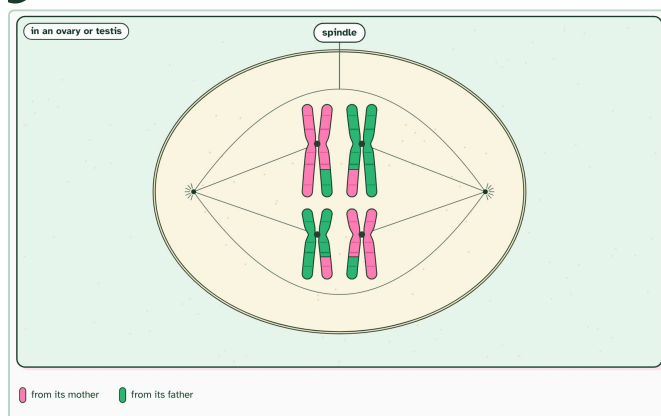


The nucleus breaks down and each chromosome lies beside its partner: a **homologous pair**, with the same genes in the same order.

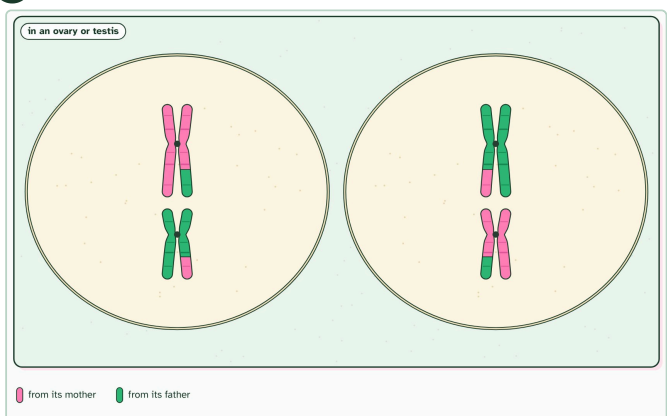


Crossing over: chromatids of a pair swap matching sections, making new mixes of the mother's and the father's alleles.

5 $2N = 4$, DNA COPIED 6 2 CELLS, $N = 2$

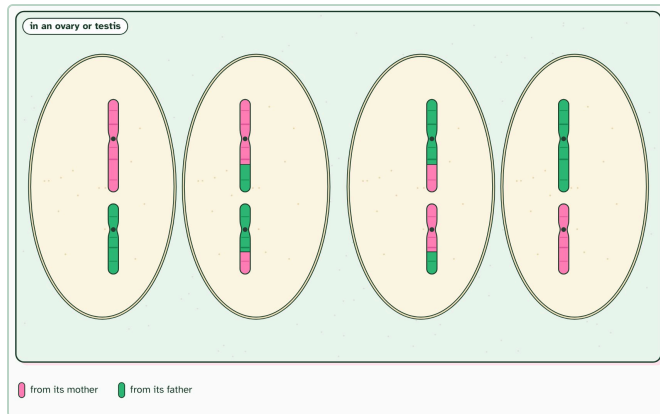


A spindle forms and the pairs line up across the middle. Which way round each pair lies is down to chance: **random orientation**.



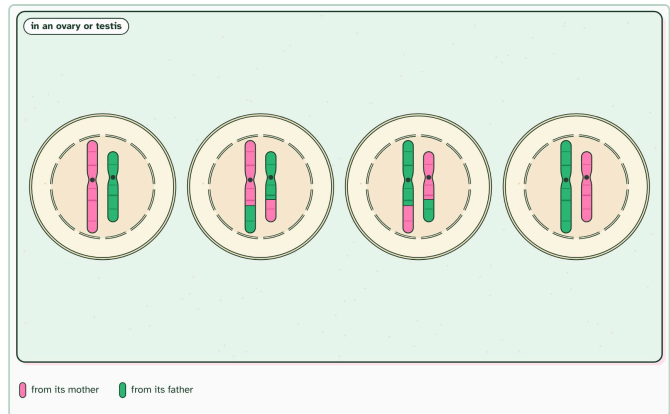
Meiosis I: one chromosome of each pair is pulled to each end, and the cell divides. Each new cell has half the number of chromosomes.

7

4 CELLS, $N = 2$ 

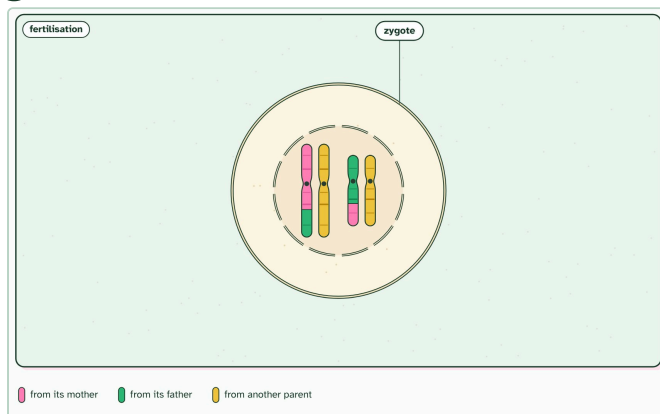
Meiosis II: in each cell the sister chromatids are pulled apart, and both cells divide again.

8

4 GAMETES, $N = 2$ 

Four **haploid** gametes, each with one chromosome of every pair, and no two alike.

9

ZYGOTE, $2N = 4$ 

Fertilisation: the gamete fuses with one from another parent (yellow). The **zygote** is diploid again, with one of each pair from each parent.

Watch it move, and change it as it runs: biologybybradford.com/guides/ib-biology-d3-2-inheritance#halve

LINKING QUESTION

What biological processes involve doubling and halving? Here it is the chromosome number: meiosis halves it and fertilisation doubles it. The same idea returns in D2.1 (cell and nuclear division), where cells divide, and in D1.1 (DNA replication), where the DNA doubles before they do.

WATCH OUT

A gamete is not "half a cell". It is a whole cell with half the number of chromosomes. And a parent does not pass both of its copies of a gene to a gamete: it passes one.

PREDICT

A human body cell has 46 chromosomes. How many chromosomes are in a sperm, and how many in the zygote it helps to form?

- (A) 46 in the sperm and 92 in the zygote, because each gamete keeps a full set
- (B) 23 in the sperm and 46 in the zygote
- (C) 23 in the sperm and 23 in the zygote, as the two sets are shared out

Decide before you look. Answer on page 52.

02

D3.2.2 GENETIC CROSSES IN FLOWERING PLANTS

How to make a *CROSS*

To study inheritance you need to know the parents, so you choose them and then count the offspring. That is a genetic cross. Flowering plants are good for it: you can control which plants breed, and the seeds are ready within weeks.

The male gametes are in **pollen**, which forms in the anthers. The female gametes are in the ovules, inside the ovary at the base of the flower. A pollen grain has to reach the stigma before it can fertilise an ovule, so a cross needs **pollination**.

Pea plants add a twist. One flower makes both pollen and ovules, so it can pollinate itself: **self-fertilisation**. To make a cross on purpose you remove the anthers from the flower that will receive pollen, before it can pollinate itself, and carry pollen from the other plant to its stigma.

Crosses are described in generations. The two parents you choose are the **P generation**. Parents that are **true-breeding** give the **F1 generation** (F is for filial, meaning of a son or daughter). Let the F1 plants pollinate each other, or themselves, and you get the **F2 generation**. A **Punnett grid** predicts what the F2 should look like. The letters in the grid are versions of a gene, which chapter 3 explains.

KEY TERMS

pollen

The tiny grains made in the anthers of a flower. Each pollen grain contains male gametes.

pollination

The transfer of pollen from an anther to a stigma.

self-fertilisation

Fertilisation of a plant's own ovules by its own pollen.

true-breeding

Plants or animals that always produce offspring like themselves when bred with their own kind, because both alleles of the gene are the same.

Punnett grid

A grid that shows every way the gametes of two parents can combine, so you can read off the genotypes of the offspring and how likely each is.

Making a cross in a pea plant

P generation



F1 generation



cross

self-pollinate

F2 generation

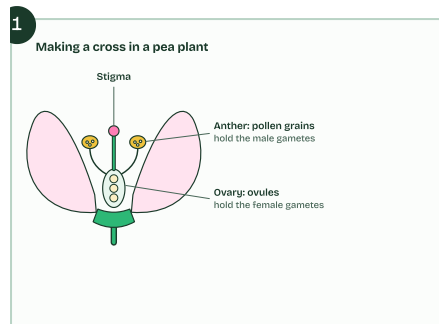
pollen from the F1 plant

	Y	y
Y	YY	Yy
y	Yy	yy

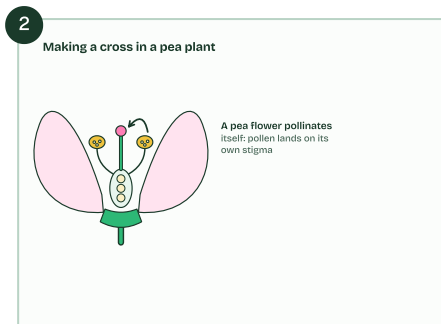
3 yellow : 1 green
1 YY : 2 Yy : 1 yy

STEP BY STEP

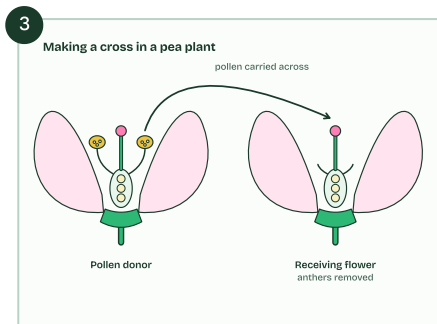
5 STEPS



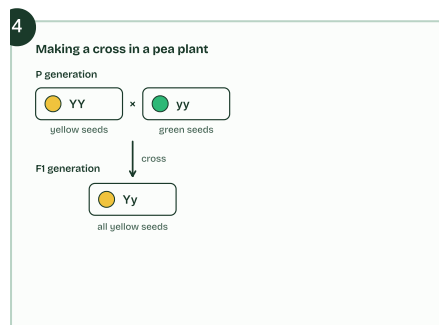
A pea flower makes both kinds of gamete. **Pollen** forms in the anthers and holds the male gametes. The **ovules** in the ovary hold the female gametes.



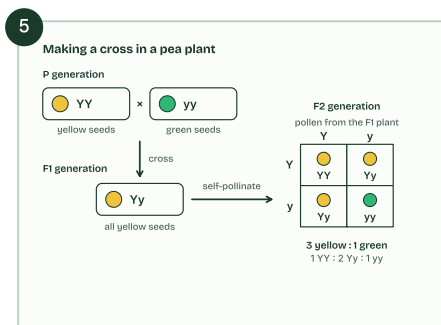
Left alone, a pea flower **pollinates itself**: its pollen reaches its own stigma.



For a **cross**, the anthers are removed from the receiving flower, and pollen is carried from the donor to its stigma.



Cross two true-breeding parents (the **P generation**). Every seed of the **F1 generation** has one allele from each parent.



Let the F1 plants self-pollinate to make the **F2 generation**. The **Punnett grid** shows every way their gametes can combine: 3 yellow seeds for every 1 green.

REMEMBER IT

The generations of a cross

P Parents
P generation the plants you choose to cross

F First
F1 generation their offspring

F Follows
F2 generation the offspring of two F1 plants

EXAMPLE

Crosses are not only for laboratories. Breeders cross plants to join the useful traits of two varieties, such as the disease resistance of one with the yield of another, and to make new varieties of ornamental flowers.

WATCH OUT

A Punnett grid gives chances, not promises. It predicts the proportions you should expect in a large number of offspring, not exactly which seeds you will get.

PREDICT

A student wants to cross a purple-flowered pea plant with a white-flowered one, but forgets to remove the anthers from the white plant. What is likely to happen?

- Ⓐ Some seeds will come from self-fertilisation, so not all will be from the cross
- Ⓑ The flower will be sterile, because a flower with both kinds of gamete cannot set seed
- Ⓒ Every seed will be purple, because pollen from another plant always beats the plant's own

Decide before you look. Answer on page 52.

03

D3.2.3 D3.2.4 GENOTYPE AND PHENOTYPE

Genes, alleles and the traits you *see*

A **gene** is a length of DNA at one place on a chromosome. Different versions of the same gene are **alleles**. The seed colour gene in peas is a single place on one chromosome, and its alleles are one for yellow seeds (Y) and one for green (y). Keep the two words apart: a gene is the place, an allele is the version.

You are diploid, so you have two alleles of every autosomal gene, one on each chromosome of a pair. The pair of alleles is your **genotype** for that gene. If the two alleles are the same, you are **homozygous** (YY or yy). If they differ, you are **heterozygous** (Yy).

Your **phenotype** is what can be observed: the colour of a seed, your blood group, your height, even your behaviour. It comes from the genotype and the environment. Some traits depend on the genotype only, some on the environment only, and many on both.

KEY TERMS

gene

A length of DNA at one place (locus) on a chromosome, with the instructions for making a polypeptide.

alleles

Alternative forms of the same gene, found at the same place on a chromosome. They differ in their base sequences.

genotype

The combination of alleles an organism has, usually for one gene.

homozygous

Having two identical alleles of a gene.

heterozygous

Having two different alleles of a gene.

phenotype

The observable traits of an organism, which result from its genotype and the environment.

Genes, alleles and genotypes

ABO blood group



a scar from a cut



adult height

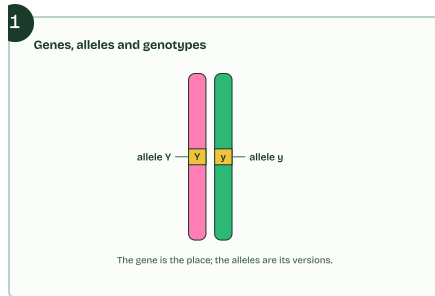


■ genotype ■ environment

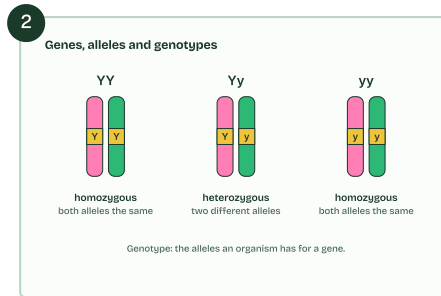
Bar lengths show the idea, not measurements.

STEP BY STEP

3 STEPS



A pair of homologous chromosomes. The **gene** is one place on the chromosome. Each chromosome carries one **allele** of it, here Y and y.



The two alleles make the **genotype**. Two the same is **homozygous**. Two different is **heterozygous**.



The **phenotype** comes from the genotype, the environment, or both: blood group, a scar and height.

EXAMPLE

In humans: **genotype only**, ABO blood group (diet and exercise cannot change it). **Environment only**, a scar from a cut, or a language you have learned. **Both**, adult height (genes set a range, and nutrition and health decide where you land in it) and skin colour (which also depends on how much sun you get).

WATCH OUT

There is no "gene for yellow seeds". There is a gene for seed colour, and the Y allele of it makes the seed yellow. A gene has alleles; a trait is made by a genotype.

PREDICT

Two plants have the same genotype for every gene, but one grows much taller because it was given more water and fertiliser. What does this show?

- (A) The taller plant must have a different set of alleles from the other
- (B) The environment affects the phenotype as well as the genotype
- (C) Fertiliser changes the alleles inside every cell of the plant

Decide before you look. Answer on page 52.

PREDICT

A pea plant has the genotype Yy. Which statement is correct?

- (A) It is homozygous, because both of its alleles belong to the same gene
- (B) It is heterozygous, because its two alleles of the gene differ
- (C) It is heterozygous, because it has two different genes for seed colour

Decide before you look. Answer on page 52.

04

D3.2.5 DOMINANT AND RECESSIVE ALLELES

Dominant and *recessive* alleles

In Mendel's peas, a cross between a yellow-seeded plant and a green-seeded one gave an F1 in which every seed was yellow. The green trait had not vanished: about a quarter of the F2 seeds were green. Something was hiding it.

A **dominant allele** shows its effect when it is present, in one copy or in two. A **recessive allele** shows only when there is no dominant allele alongside it. So YY and Yy have the same phenotype, yellow, and only yy is green. By convention, a capital letter is the dominant allele and a small letter the recessive one.

Why is one copy enough? An allele works through the protein its gene makes. For many genes, a recessive allele is a version that does not make a working protein. A heterozygote has one working copy, and for many genes one working copy makes enough protein to give the trait. A homozygous recessive has no working copy, and the trait changes. There is no single mechanism for all genes, but this is the usual reason.

KEY TERMS

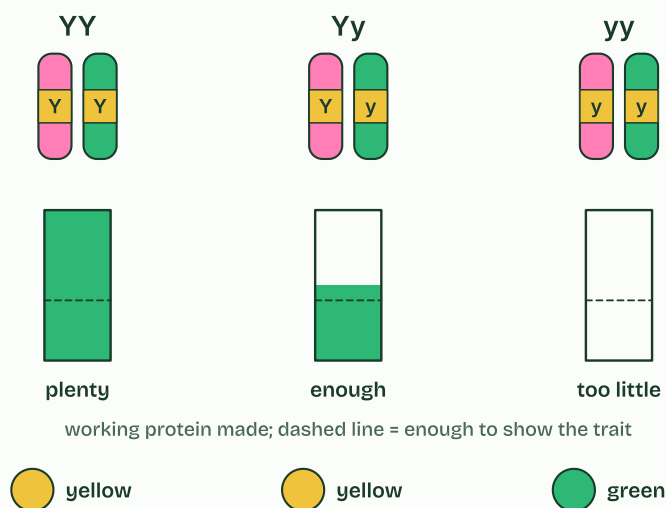
dominant allele

An allele whose effect shows in the phenotype of a heterozygote as well as of a homozygote.

recessive allele

An allele whose effect shows only when there is no dominant allele, so the organism is homozygous for it.

Three genotypes of one gene



STEP BY STEP

1 Three genotypes of one gene

Three genotypes of the seed colour gene: two **homozygous** and one **heterozygous**.

2 Three genotypes of one gene

working protein made; dashed line = enough to show the trait

Each gene makes a protein. A **Y** allele makes a working one, and one working copy makes **enough** to give the trait. With no working copy, the trait changes.

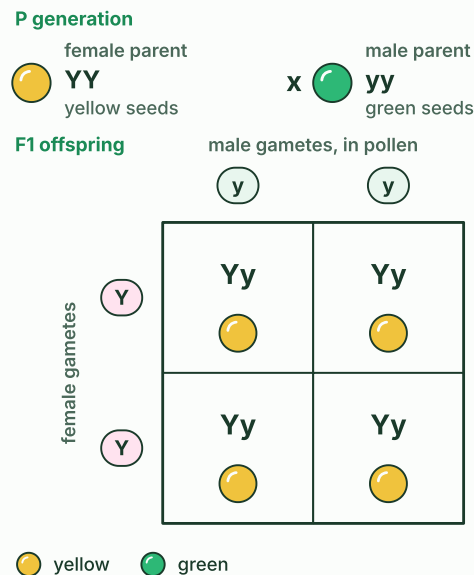
3 STEPS

3 Three genotypes of one gene

working protein made; dashed line = enough to show the trait

yellow yellow green

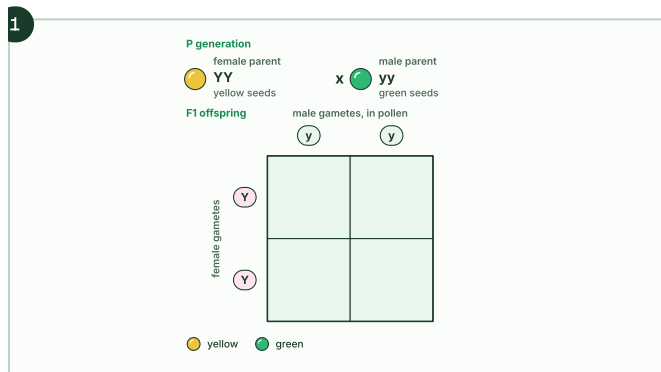
The phenotypes: **YY** and **Yy** are both yellow, so the dominant allele shows in a heterozygote. Only **yy** is green.



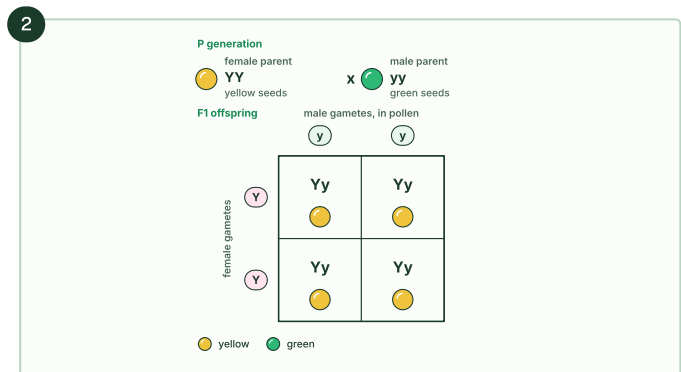
Pick the two parents' genotypes, predict the offspring, then breed 30 and see how the sample scatters around the expectation.

STEP BY STEP

2 STEPS



Each parent makes gametes by meiosis, one allele of each gene in each. The female parent's go down the side of the **Punnett grid**, the male parent's along the top.



Each cell joins one gamete from each parent: a possible offspring, every cell as likely as the next. The F1: genotypes all Yy; phenotypes all yellow.

WATCH OUT

Dominant does not mean common, strong or better. Some dominant alleles are rare, and some cause disease. Dominant only describes what shows in a heterozygote. Recessive alleles do not fade away either: in a heterozygote they are still there, and they can reappear in a later generation.

PREDICT

Two yellow-seeded pea plants are crossed, and some of their offspring have green seeds. What must be true of both parents?

- (A) Both are homozygous YY, and the green seeds arose by chance
- (B) Both are heterozygous Yy: each carries a hidden y allele
- (C) One is YY and the other is Yy, so a green seed can appear

Decide before you look. Answer on page 52.

PREDICT

Two Yy plants are crossed. What fraction of their offspring have green seeds?

- ☐ (A) One half
- ☐ (B) Three quarters
- ☐ (C) One quarter

Decide before you look. Answer on page 52.

05

D3.2.6 PHENOTYPIC PLASTICITY

Same genes, *different outcomes*

Genotype is not destiny. One individual, with one set of alleles, can develop different traits in different conditions, and the change often suits the conditions. That is **phenotypic plasticity**.

Take two cuttings from one plant. They have identical alleles. Grow one in full sun and the other in shade. A shaded plant tends to grow taller, with longer gaps between its leaves and broader leaves, reaching for light. The sunlit plant stays shorter and sturdier. Neither plant changed its genotype.

The difference is in **gene expression**: which genes are switched on, where, and how strongly. Light conditions change the pattern, so the same DNA builds a different body. The base sequence has not changed, which means plasticity is not a mutation.

Because the alleles are the same, the change can be **reversible** during the life of the individual: move the shaded plant into the sun, and its new growth comes in short and sturdy, like the sun plant's. Growth that has already happened stays as it was.

KEY TERMS

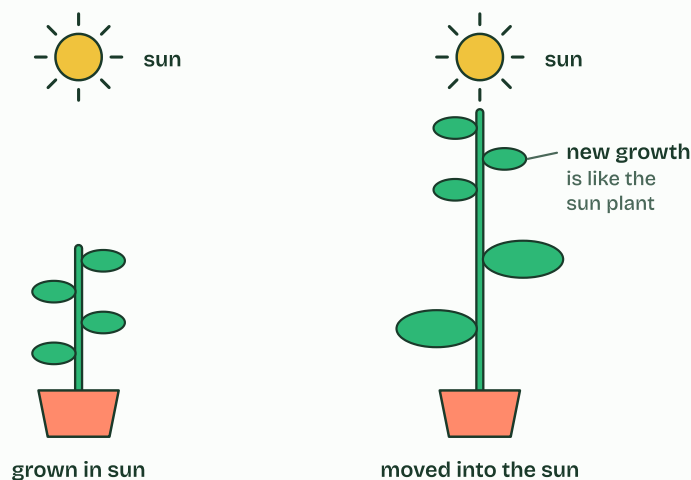
phenotypic plasticity

The ability of one genotype to produce different phenotypes, by changing which genes are expressed, in response to the environment the organism experiences.

gene expression

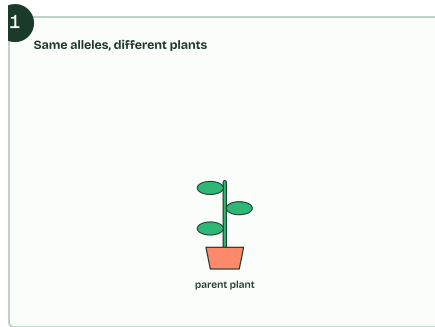
The use of the information in a gene to make a product, so that the gene affects the phenotype.

Same alleles, different plants

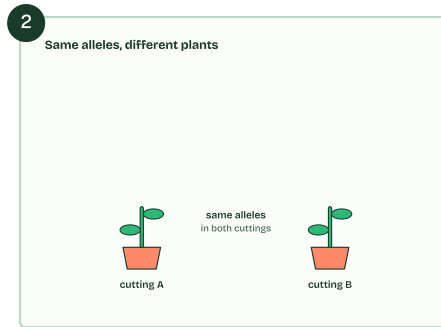


STEP BY STEP

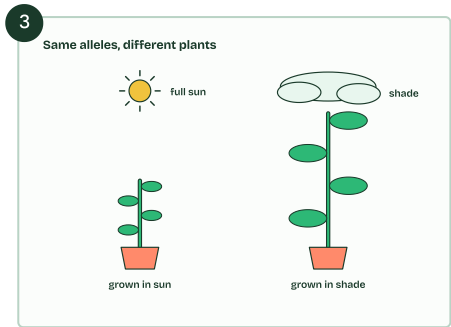
4 STEPS



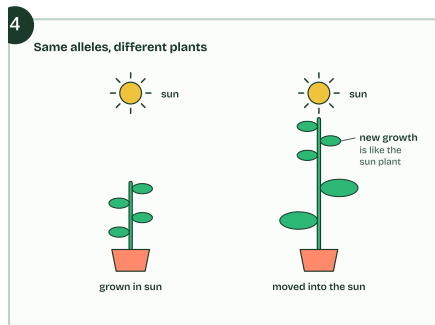
One parent plant, with its alleles.



Two **cuttings** from it carry exactly the same alleles.



One cutting grows in **sun** and one in **shade**. Same alleles, but different gene expression makes a short, sturdy plant and a tall one with broad leaves.



Move the shaded plant into the sun, and its **new growth** comes in like the sun plant's. The change is reversible, and the alleles never changed.

EXAMPLE

Your skin tans in strong sun and fades again afterwards, with no change in your DNA. An Arctic hare's coat is white in winter and darker in summer, so the same animal is camouflaged in each season.

WATCH OUT

A plastic change is not passed to the next generation through the alleles: the shaded plant's seeds do not inherit its stretching. The individual does not choose to change either. The capacity to respond is itself in the genotype, and the environment triggers it.

PREDICT

Two clones grow differently, one in sun and one in shade. What explains the difference?

- (A) One seedling has mutated and now has different alleles
- (B) The light changed how their genes were expressed
- (C) The shaded seedling lost the alleles for sturdy growth

Decide before you look. Answer on page 52.

PREDICT

Which of these is an example of phenotypic plasticity?

- (A) A baby has the same ABO blood group as her father
- (B) A beetle population grows darker over many generations of selection
- (C) A person's skin tans in strong sun and fades in winter

Decide before you look. Answer on page 52.

06

D3.2.7 PHENYLKETONURIA: A RECESSIVE DISEASE

One faulty enzyme: *PKU*

Phenylketonuria (PKU) is a human disease caused by a recessive allele of an autosomal gene. The gene codes for an enzyme, phenylalanine hydroxylase, which converts the amino acid phenylalanine into another amino acid, tyrosine.

A person with at least one working allele makes enough enzyme. A person who is homozygous for the faulty allele makes none that works. Phenylalanine from food cannot be converted, so it builds up, and in a baby the high levels can damage the developing brain.

Because the allele is recessive, a person with one working and one faulty allele has no symptoms. That person is a **carrier**. Two carrier parents have a 1 in 4 chance of a child with PKU each time they have a baby, and most carriers do not know that they are one.

PKU shows genotype and environment working together. The genotype cannot be changed, but the environment can. Babies are screened soon after birth, and a child found to have PKU is given a diet low in phenylalanine, which protects the developing brain. The phenotype depends on the diet as well as on the alleles.

KEY TERMS

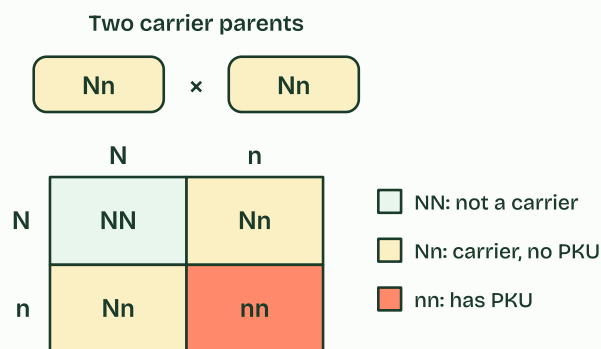
phenylketonuria

A recessive genetic condition in which the body cannot convert phenylalanine to tyrosine, because the enzyme that does this is not working.

carrier

Someone who has one copy of a recessive allele for a condition, is not affected by it, and can pass the allele on.

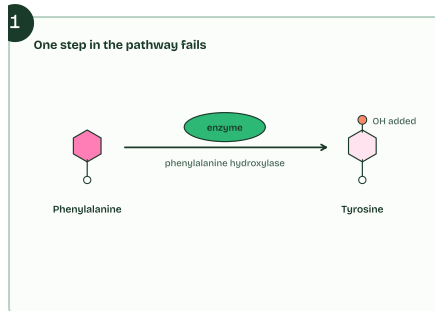
One step in the pathway fails



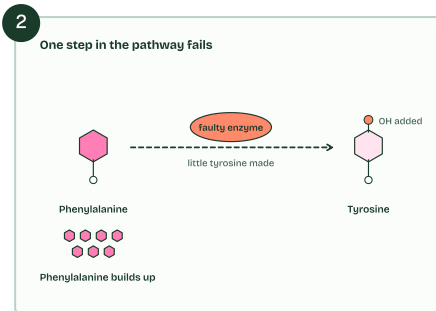
Each child has a 1 in 4 chance of PKU.

STEP BY STEP

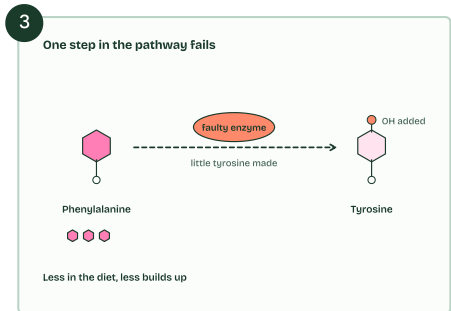
4 STEPS



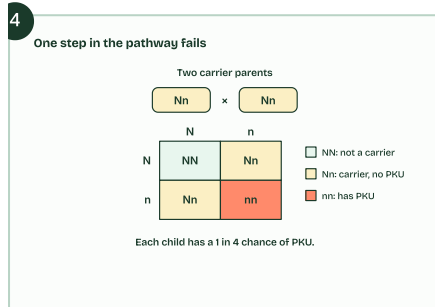
In a person with a working allele, the **enzyme** converts phenylalanine to tyrosine, which has one extra OH group.



In a person with two faulty alleles the enzyme does not work. Little tyrosine is made and **phenylalanine builds up**.



A **diet low in phenylalanine** gives the body less to build up. The genotype is the same, and the phenotype changes.



Two **carrier** parents (Nn): each child has a 1 in 4 chance of nn, so of PKU.

EXAMPLE

Two carriers, $Nn \times Nn$. Each makes N and n gametes in equal numbers. The offspring are NN (1 in 4), Nn (2 in 4) and nn (1 in 4). Only nn has PKU. Of the children who do not have PKU, two in three are carriers.

WATCH OUT

The chance is 1 in 4 for each child, every time. One child with PKU does not make the next three safe.

PREDICT

Two parents do not have PKU, but their first child does. What is the chance that their second child has PKU?

- (A) 1 in 2, because one of the two parents must be affected
- (B) None, because the first child used up the recessive alleles
- (C) 1 in 4, because both parents must be carriers

Decide before you look. Answer on page 53.

PREDICT

A baby with PKU is put on a diet low in phenylalanine. Has its genotype changed?

- (A) Yes, the diet slowly repairs the faulty allele in its cells
- (B) No: the genotype is the same, but the phenotype changes
- (C) Yes, but only in the liver, where the enzyme is normally made

Decide before you look. Answer on page 53.

07

D3.2.8 D3.2.9 SNPS, MULTIPLE ALLELES AND ABO BLOOD GROUPS

Many alleles, not just two

So far every gene had two alleles. In a population, a gene can have many. New alleles arise by mutation, and alleles of one gene usually differ by only one or a few bases. A place in the genome where people differ by a single base, and where each version is common enough to count (often taken as more than 1 in 100), is a **single-nucleotide polymorphism**, or SNP ("snip").

All the alleles of all the genes in a population make up its **gene pool**. A gene can have any number of alleles in the gene pool, but you are diploid, so you inherit only two of them: one from each parent. Different people carry different pairs.

The **ABO blood group** is the textbook example. One gene has three alleles: I^A , I^B and i . I^A and I^B each lead to a different marker (an antigen) on the surface of red blood cells, and i leads to neither. Both I^A and I^B are dominant to i . Three alleles make six genotypes and four blood groups; what happens when I^A meets I^B is the subject of chapter 8.

KEY TERMS

single-nucleotide polymorphism

Single-nucleotide polymorphism: a position in the genome where individuals differ by one base, and where each version is common in the population.

gene pool

All the alleles of all the genes in a population.

ABO blood group

The system of human blood groups A, B, AB and O, controlled by one gene with three alleles.

SNPs, alleles and the gene pool

genotype	blood group
$I^A I^A$	group A
$I^A i$	group A
$I^B I^B$	group B
$I^B i$	group B
$I^A I^B$	group AB
ii	group O

Three alleles in the pool, so six genotypes and four groups.

STEP BY STEP

4 STEPS

1 SNPs, alleles and the gene pool

Person 1: A T G G C A C T G A C T

Person 2: A T G G C A C T G A C T

Person 3: A T G G C A T T G A C T

Person 4: A T G G C A C T G A C T

A SNP: one position where people differ by one base

DNA from four people at one place. They differ at a single base: a **SNP**.

2 SNPs, alleles and the gene pool

Gene pool

one gene, four alleles in the population

Across a whole population a gene can have many **alleles**. The **gene pool** holds them all.

3 SNPs, alleles and the gene pool

Gene pool

one gene, four alleles in the population

→

One person inherits just two

Each person inherits just **two** of them.

4 SNPs, alleles and the gene pool

genotype	blood group
$I^A I^A$	group A
$I^A i$	group A
$I^B I^B$	group B
$I^B i$	group B
$I^A I^B$	group AB
ii	group O

Three alleles in the pool, so six genotypes and four groups.

ABO: three alleles in the pool give six genotypes and four blood groups.

REMEMBER IT

The four blood groups

A Apple Group A $I^A I^A$ or $I^A i$	B Banana Group B $I^B I^B$ or $I^B i$	B Both Group AB $I^A I^B$, with both markers	O Orange Group O ii, with neither marker
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EXAMPLE

A parent of group A ($I^A i$) and a parent of group B ($I^B i$) can have children of any of the four groups: $I^A I^B$ (AB), $I^A i$ (A), $I^B i$ (B) or ii (O).

WATCH OUT

A person never has three ABO alleles. The gene pool has three; each person has two. And group O is not "no gene": the i allele is a real allele, and a person with ii has two copies of it.

PREDICT

A man with blood group AB ($I^A I^B$) and a woman with group O (ii) have children. Which groups are possible?

- (A) A, B, AB and O
- (B) AB and O only
- (C) A and B only

Decide before you look. Answer on page 53.

PREDICT

How many alleles of the ABO gene does one person have?

- (A) Three, one for each allele in the gene pool
- (B) Two, one from each parent
- (C) One, because the gene is on one chromosome

Decide before you look. Answer on page 53.

08

D3.2.10 INCOMPLETE DOMINANCE AND CODOMINANCE

Neither wins, *or both do*

Dominance is not the only pattern. Sometimes a heterozygote looks like neither homozygote. Two patterns are worth telling apart, and the difference is in the phenotype.

In **incomplete dominance** the heterozygote has an **intermediate** phenotype. The standard example is the four o'clock plant, *Mirabilis jalapa*, also called the marvel of Peru. The alleles are written C^R (red) and C^W (white). $C^R C^R$ plants are red, $C^W C^W$ white and $C^R C^W$ pink. Cross two pink plants and you get red, pink and white in the ratio 1 : 2 : 1, so here the phenotype ratio matches the genotype ratio.

In **codominance** both alleles are expressed fully, and the heterozygote has a **dual** phenotype. The AB blood group is the example: a person with $I^A I^B$ makes both the A marker and the B marker on every red blood cell. Neither allele hides the other, and nothing is in between: both are there.

KEY POINTS

- Complete dominance: the heterozygote looks like one homozygote.
- Incomplete dominance: the heterozygote is in between.
- Codominance: the heterozygote shows both at once.

KEY TERMS

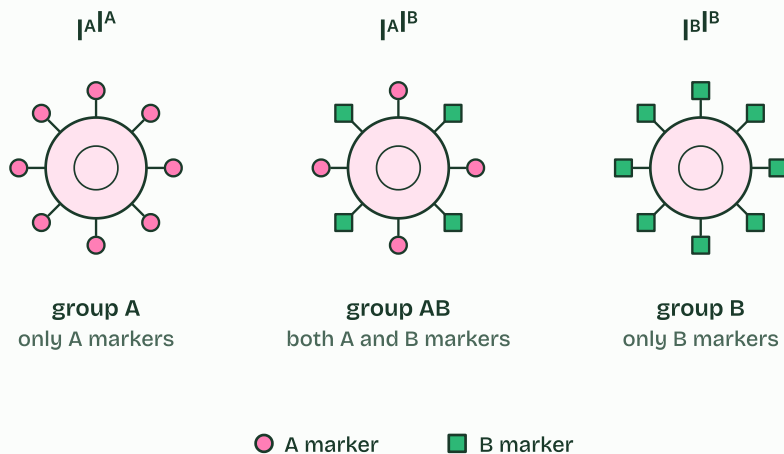
incomplete dominance

A pattern of inheritance in which the heterozygote has a phenotype in between those of the two homozygotes.

codominance

A pattern of inheritance in which both alleles of a heterozygote are fully expressed, giving a dual phenotype.

When neither allele wins outright



STEP BY STEP

3 STEPS

1 When neither allele wins outright

$C^R C^R$ $C^R C^W$ $C^W C^W$

red homozygous pink heterozygous white homozygous

Four o'clock flowers (*Mirabilis jalapa*)

Incomplete dominance in four o'clocks: red ($C^R C^R$), pink ($C^R C^W$) and white ($C^W C^W$). The heterozygote is intermediate.

2 When neither allele wins outright

pink × pink

	C^R	C^W
C^R	$C^R C^R$	$C^R C^W$
C^W	$C^R C^W$	$C^W C^W$

1 red : 2 pink : 1 white
The phenotype ratio matches the genotype ratio.

Cross two pink plants and the offspring are **1 red : 2 pink : 1 white**. The phenotype ratio matches the genotype ratio, and red and white reappear.

3 When neither allele wins outright

$I^A I^A$ $I^A I^B$ $I^B I^B$

group A only A markers group AB both A and B markers group B only B markers

● A marker ■ B marker

Codominance in blood: a red blood cell of group AB carries both markers, so the heterozygote has a dual phenotype.

WATCH OUT

Incomplete dominance is not blending. If the red and white alleles mixed like paints, pink plants could never give red or white again. They do, in the F₂, because each allele stays whole and separates in the gametes.

PREDICT

A red four o'clock plant is crossed with a white one. What colour are the F₁ flowers?

- (A) All red, since red is dominant
(B) Half red and half white
(C) All pink

Decide before you look. Answer on page 53.

PREDICT

What blood group does a person with the genotype $I^A I^B$ have, and why?

- (A) Group A, because I^A is stronger than I^B
(B) Group AB, because both alleles are expressed
(C) Group O, because the two alleles cancel each other out

Decide before you look. Answer on page 53.

09

D3.2.11 D3.2.12 SEX DETERMINATION AND SEX LINKAGE

X, Y and *sex*

Most of your 23 pairs of chromosomes are autosomes, matching pairs. The last pair is different. Human females have two X chromosomes (XX). Males have one X and a smaller Y (XY). Every egg carries an X. A sperm carries an X or a Y, so the sperm decides: an X sperm makes an XX zygote, which develops female-typical characteristics, and a Y sperm makes an XY zygote, which develops male-typical ones. Half the sperm carry each, so about half of children are girls. The Y chromosome carries genes that start the development of male characteristics.

The X and the Y are very unequal. The X carries far more genes than the Y, and most of them have nothing to do with sex. A **sex-linked** gene is one that sits on a sex chromosome, and in practice nearly all of them are on the X. A male has only one X, so a recessive allele on it always shows: there is no second allele on the Y to mask it. A female has two Xs and can be a carrier.

Haemophilia is the standard example. A recessive allele on the X means a protein needed to clot blood is missing or faulty, so even small cuts can bleed for a long time. Write the alleles as superscripts on an uppercase X: X^H (normal clotting) and X^h (haemophilia). A girl $X^H X^h$ is a carrier. A boy $X^h Y$ has haemophilia, because he has no other X. The Y gets no letter, because it has no allele of this gene.

KEY TERMS

sex-linked

A gene on a sex chromosome, so its pattern of inheritance differs between males and females.

haemophilia

An X-linked recessive disorder in which blood clots slowly or not at all, because a protein needed for clotting is missing or does not work.

X, Y and sex

carrier mother × unaffected father

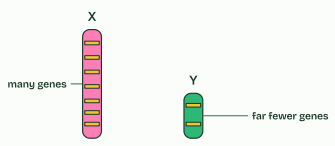
	X^H	Y
X^H	$X^H X^H$ girl, not affected	$X^H Y$ boy, not affected
X^h	$X^H X^h$ girl, carrier	$X^h Y$ boy, has haemophilia

Mother: row of eggs. Father: column of sperm.

STEP BY STEP

3 STEPS

1 X, Y and sex



many genes

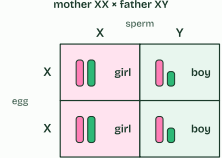
far fewer genes

Two X chromosomes: female characteristics develop.
One X and one Y: male characteristics develop.

The **X** is big and carries many genes. The **Y** is small and carries far fewer. Two Xs develop female, an X and a Y develop male.

2 X, Y and sex

mother XX × father XY



Half the sperm carry X and half carry Y: the sperm decides.

Every egg carries an X. A sperm carries X or Y, so the **sperm decides** the sex: half girls, half boys.

3 X, Y and sex

carrier mother × unaffected father

	X^H	Y
X^H	$X^H X^H$ girl, not affected	$X^H Y$ boy, not affected
X^h	$X^H X^h$ girl, carrier	$X^h Y$ boy, has haemophilia

Mother: row of eggs. Father: column of sperm.

Haemophilia: a carrier mother ($X^H X^h$) and an unaffected father ($X^H Y$). Half the sons have haemophilia and half the daughters are carriers. No daughter is affected.

EXAMPLE

Red-green colour blindness is another X-linked recessive condition, so it is more common in males than in females.

WATCH OUT

A father cannot pass an X-linked allele to his son. A son gets his Y from his father and his X from his mother. A father does pass his X to every daughter.

PREDICT

A man with haemophilia ($X^h Y$) and a woman who is not a carrier ($X^H X^H$) have children. What are the children like?

- (A) All sons have haemophilia; all daughters are unaffected
- (B) Half the sons and half the daughters have haemophilia
- (C) All daughters are carriers; all sons are unaffected

Decide before you look. Answer on page 53.

PREDICT

Why are X-linked recessive conditions more common in males than in females?

- (A) The Y chromosome makes the faulty protein in males
- (B) Males receive their X chromosome from their father
- (C) Males have one X, so one recessive allele shows

Decide before you look. Answer on page 53.

10

Reading a *family tree*

D3.2.13 PEDIGREE CHARTS

A **pedigree chart** follows a trait through a family. Squares are males and circles are females. A filled shape means the person has the condition, and a line across joins a couple, with their children below. Generations are numbered I, II, III from the top.

You cannot cross people to see what happens, so you read the pattern instead. The aim is to work out whether the condition is dominant or recessive, and whether the gene is autosomal or on the X. Some patterns settle it. Others leave more than one possibility.

Chart reading uses two kinds of reasoning. Looking over the family and deciding "this pattern fits a recessive trait" is **inductive reasoning**: you generalise from the cases you can see. Once you have the pattern, you use **deductive reasoning** to find the genotype of a particular person: if the trait is recessive, an affected woman must be nn , and her unaffected parents must both be Nn .

Pedigrees also explain a rule found in many societies, that close relatives may not marry. Relatives share alleles they inherited from common ancestors. If a harmful recessive allele runs in a family, two relatives are more likely to carry the same one, so their children are more likely to be homozygous recessive and have the condition. That is the genetic basis only; the rule has other roots too.

KEY TERMS

pedigree chart

A chart of the members of a family over several generations, showing who is affected by a genetic condition.

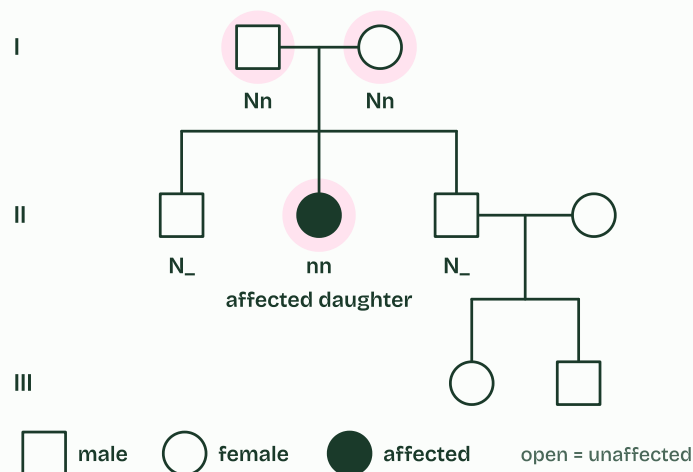
inductive reasoning

Reasoning from observations of some cases to a general conclusion or theory. The conclusion is probable, not certain.

deductive reasoning

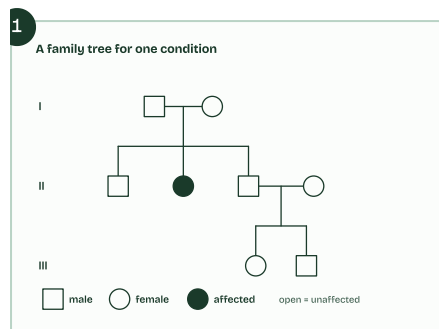
Reasoning from a general rule to a conclusion about a particular case. If the rule and the facts are true, the conclusion is certain.

A family tree for one condition

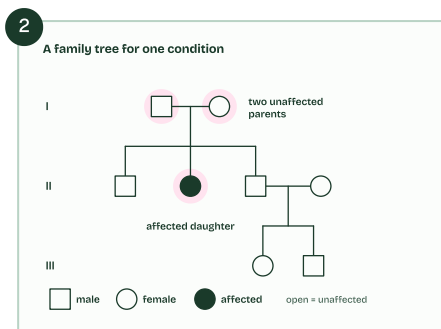


STEP BY STEP

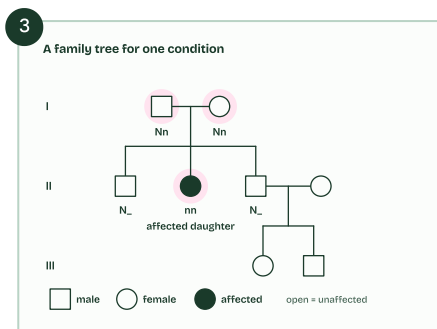
3 STEPS



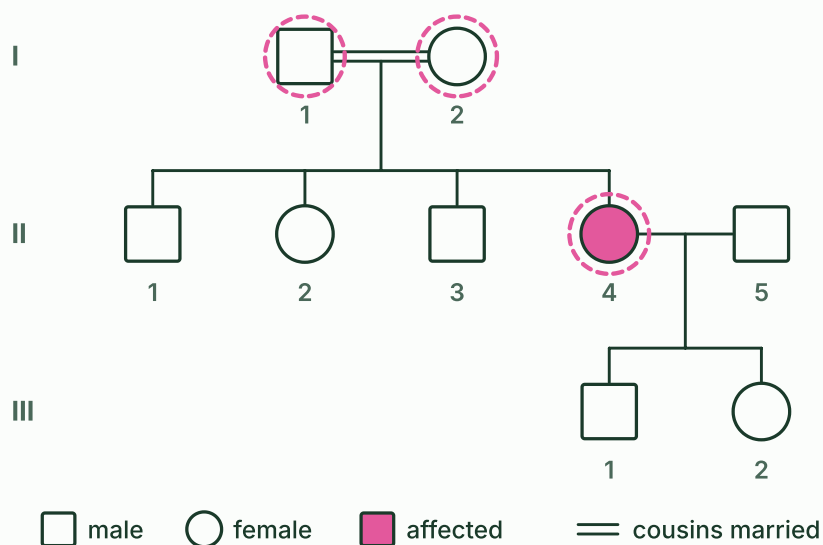
The key: **squares** are males, **circles** are females, filled symbols are affected. A line across joins a couple, and lines down lead to their children.



Two unaffected parents (generation I) have an affected daughter (generation II). The condition was **hidden** in both parents.



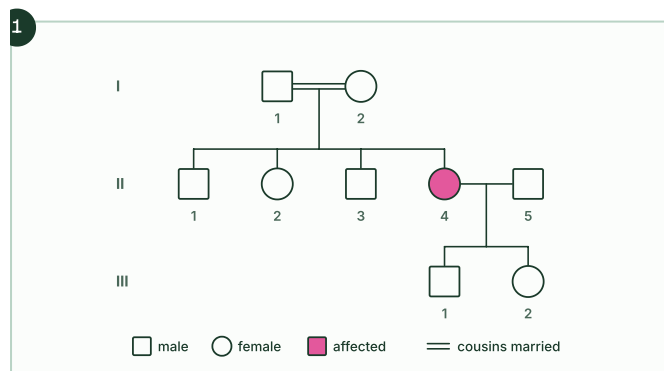
Deducing the genotypes: a recessive condition needs **nn** in the daughter and one **n** from each parent, so both parents are **Nn**.



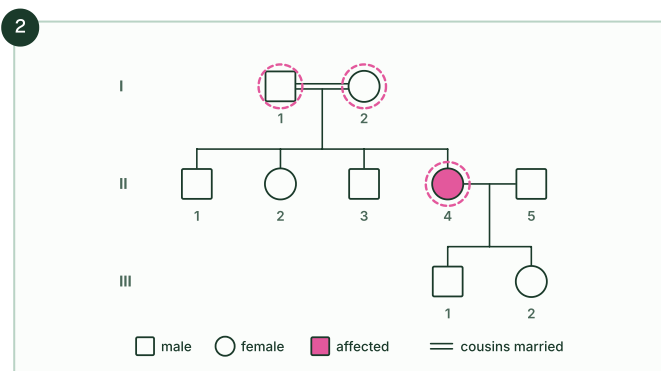
Mark who is affected, then test four ideas (autosomal dominant, autosomal recessive, X-linked recessive, X-linked dominant) and see which the family rules out, and why.

STEP BY STEP

2 STEPS



A condition no one in the family had heard of appears in a daughter. Her parents are first cousins. Squares are men, circles women; a filled symbol is **affected**. Roman numerals number the generations.



Look first at I-1, I-2 and II-4, ringed: they rule out at least one way of inheriting. Which patterns could give this tree, and which are **ruled out**?

REMEMBER IT

Four clues for reading a pedigree

Q Quiet	L Loud	D Dad	B Boys
Unaffected parents, affected child recessive: both parents carry the allele	Affected parents, unaffected child dominant: both parents carry a recessive allele	Affected daughter, unaffected father not X-linked recessive	Mostly sons, skipping generations suggests X-linked recessive

WATCH OUT

A pattern that fits is not proof. A small family may fit more than one inheritance pattern, so say "consistent with" unless the chart rules the others out.

PREDICT

In a pedigree, two unaffected parents have an affected daughter. Which statement is correct?

- (A) It is dominant, because it appeared in the child and in the family
- (B) It is X-linked dominant, because the affected child is female
- (C) It is recessive and autosomal, so both parents carry it

Decide before you look. Answer on page 54.

PREDICT

You decide, from several families, that a trait is probably recessive. What kind of reasoning is this?

- (A) Deductive: it starts from a rule and applies it to cases
- (B) Inductive: it generalises from the families seen
- (C) Deductive: the conclusion is certain whenever it is made

Decide before you look. Answer on page 54.

NATURE OF SCIENCE

Scientists draw general conclusions by inductive reasoning: they base a theory on some of the cases, not all. A pattern deduced from part of a pedigree is a theory, and it then lets you deduce the genotypes of particular people. Keep the two kinds of reasoning apart in your answers.

11

D3.2.14 CONTINUOUS VARIATION

A spread, *not a few types*

Some traits come in a few clear types. ABO blood group is A, B, AB or O, and nobody is halfway. That is **discrete variation**, and it usually comes from one gene with a few alleles. Other traits, such as height, skin colour or the mass of a seed, form a smooth range with every value in between. That is **continuous variation**.

What makes the difference? Two things, often together. A trait can be **polygenic**: controlled by several genes whose small effects add up. With one gene you get a few types. With many genes you get many combinations of alleles and a smooth spread. And the **environment** blurs the steps further, because it nudges every genotype a little up or down.

Human skin colour is the standard example. The amount of the pigment melanin depends on several genes, and also on sun exposure. The figure uses a very simple model: six alleles, each making the skin a little lighter or a little darker, like six coins. Most combinations land near the middle, and the extremes are rare. The real picture has more genes with unequal effects, but the pattern is the same: a smooth spread with most people near the middle.

You describe the middle of such data with three numbers. The **mean** is the sum divided by the number of values. The **median** is the middle value when they are in order. The **mode** is the most frequent value.

KEY TERMS

discrete variation

Variation in which individuals fall into separate categories with nothing in between, such as blood group.

continuous variation

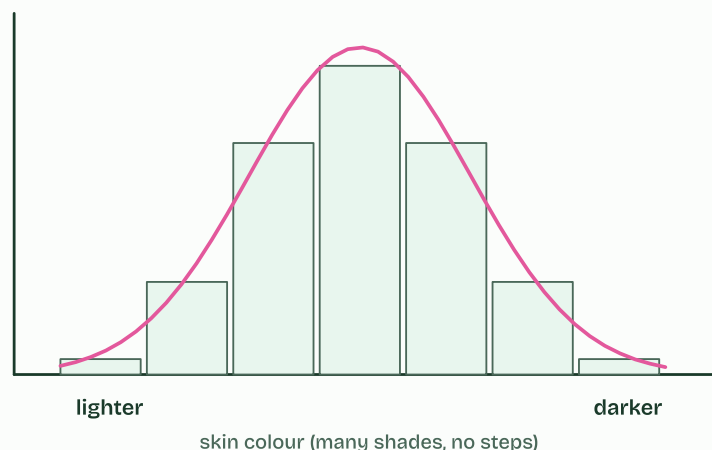
Variation in which individuals can have any value across a range, with no separate categories, such as height or skin colour.

polygenic

Controlled by several genes, each adding a small effect.

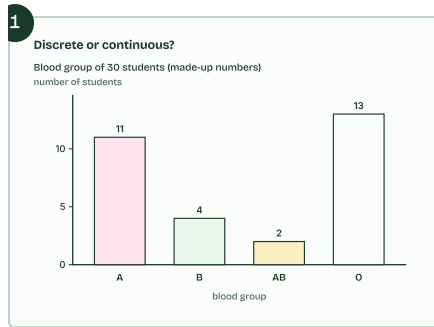
Discrete or continuous?

Add sun, diet and other environmental effects

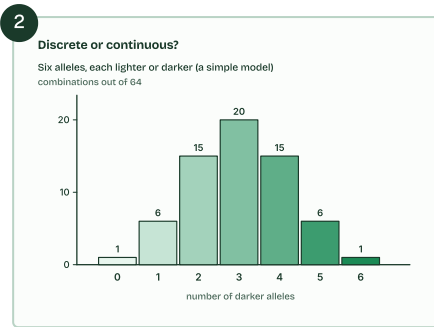


STEP BY STEP

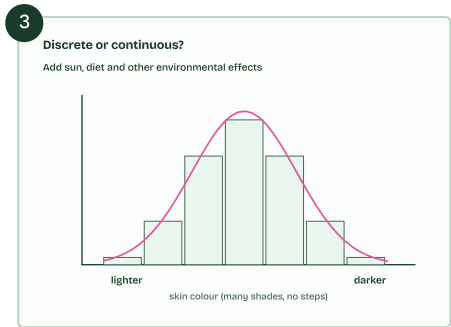
3 STEPS



Discrete variation: the bars are separate, because nobody is halfway between two blood groups.



A simple model of a **polygenic** trait: six alleles, each lighter or darker. Combinations with three darker alleles are commonest, and the extremes are rare.



Add the **environment** and the steps blur into a smooth, **continuous** spread.

EXAMPLE

Five heights in centimetres: 150, 158, 160, 160 and 182. The mean is $810 \div 5 = 162$, the median is 160 and the mode is 160. One tall value pulls the mean up but leaves the median where it was.

WATCH OUT

Continuous does not mean "not genetic". Height is strongly affected by genes. And polygenic does not mean "no dominance": each of the genes can still have dominant and recessive alleles.

PREDICT

Which of these shows discrete variation?

- (A) The heights of all the students in a class
- (B) The blood groups of students in a class
- (C) The masses of seeds taken from one plant

Decide before you look. Answer on page 54.

12

D3.2.15 BOX-AND-WHISKER PLOTS

Showing a spread with a *box plot*

A graph of continuous data should show the middle and the spread. A **box-and-whisker plot** does it with six aspects of the data: the minimum, the first quartile, the median, the third quartile, the maximum and any outliers.

Put the values in order. The median splits them in half. The **quartiles** split each half again: a quarter of the values are below Q_1 and a quarter are above Q_3 . The box runs from Q_1 to Q_3 with the median marked inside, so half of all the data are in the box. The length of the box, $Q_3 - Q_1$, is the **interquartile range** (IQR). Calculators and spreadsheets work out quartiles by slightly different rules, so use the rule you are given.

A value is an **outlier** if it is more than $1.5 \times \text{IQR}$ above Q_3 or below Q_1 . In the figure the IQR is $172 - 160 = 12$, so the fences are at $160 - 18 = 142$ and $172 + 18 = 190$. The whiskers run to the lowest and highest values inside the fences, 150 and 181, and the 194 cm student is plotted on their own.

KEY TERMS

box-and-whisker plot

A graph that shows the minimum, first quartile, median, third quartile and maximum of a data set as a box with two whiskers, and any outliers as separate points.

quartiles

The values that split ordered data into four equal groups. A quarter of the data lie below the first quartile (Q_1) and a quarter above the third (Q_3).

interquartile range

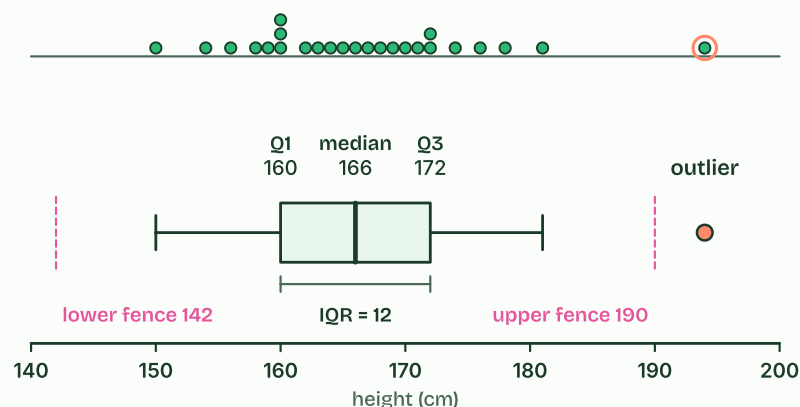
Interquartile range: Q_3 minus Q_1 , the spread of the middle half of the data.

outlier

A data point more than 1.5 times the IQR above the third quartile or below the first quartile.

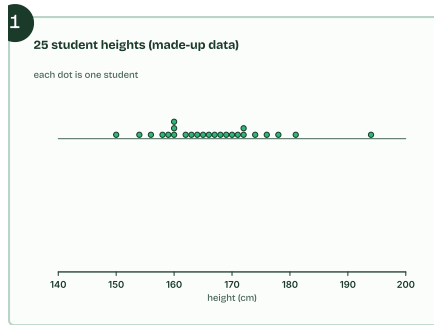
25 student heights (made-up data)

each dot is one student

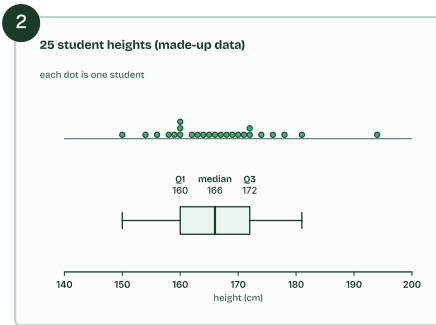


STEP BY STEP

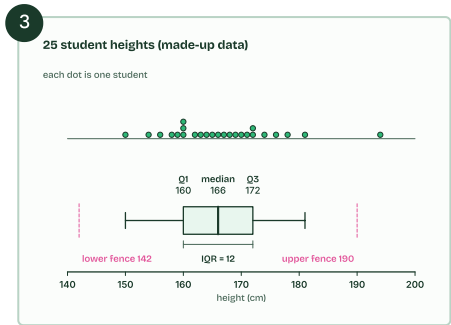
4 STEPS



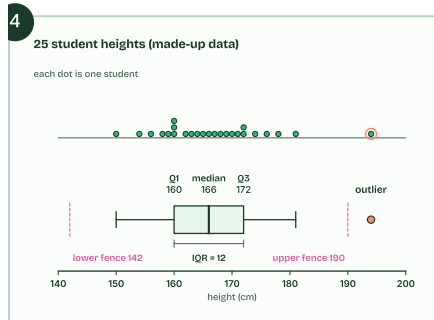
25 student heights, one dot each (made up). Where is the middle, and how spread out are they?



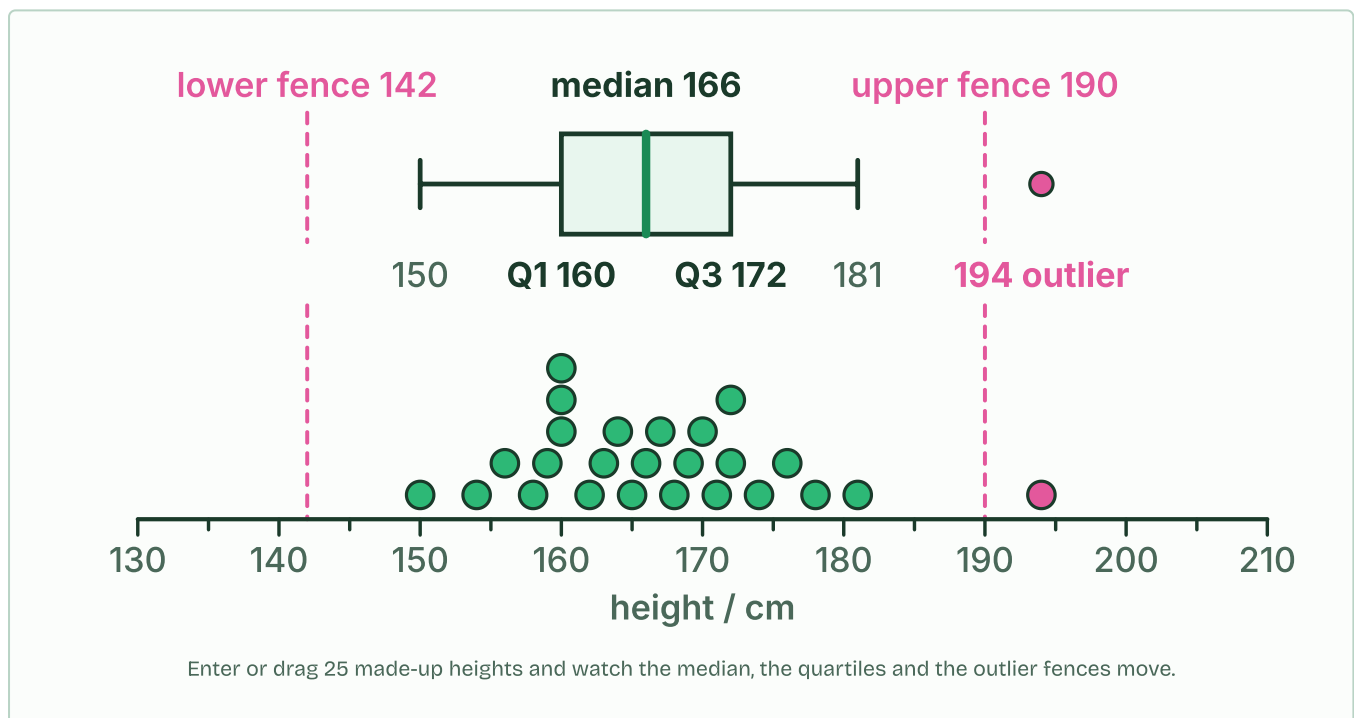
The box runs from **Q1** to **Q3** with the **median** marked. The whiskers run to the smallest and largest values that are not outliers.



IQR = $172 - 160 = 12$. The fences lie $1.5 \times \text{IQR}$ beyond the box, at 142 and 190.

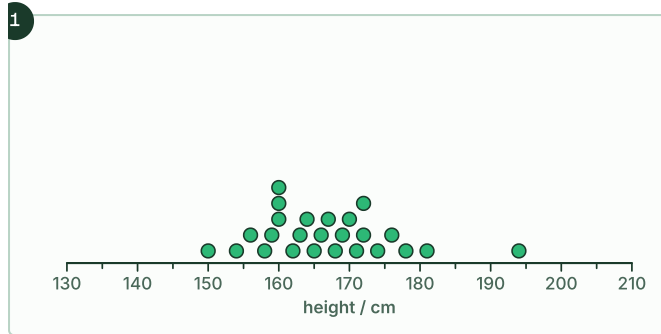


A value beyond a fence is an **outlier**: 194 is above 190, so it is plotted on its own.



STEP BY STEP

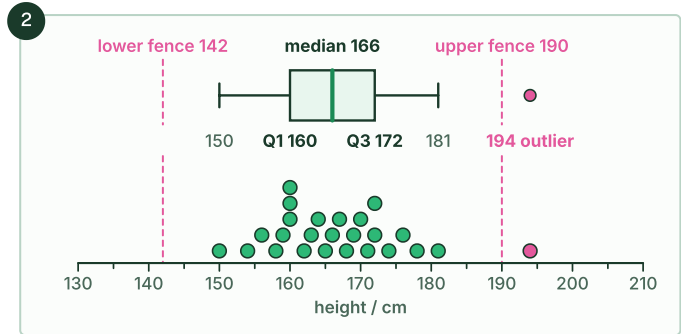
2 STEPS



Heights of 25 students in one class (made-up data). Height varies **continuously**: each dot is one student, and the values spread out with most near the middle.

EXAMPLE

For the 25 made-up heights: median 166 cm, mean 166.8 cm (to one decimal place), mode 160 cm and range 44 cm (150 to 194). The one outlier pulls the mean a little above the median.



The **box** runs from Q_1 (160) to Q_3 (172) with the **median** (166) inside; the whiskers reach the furthest values within $1.5 \times IQR$ of the box (the dashed fences, 142 and 190). Beyond them, 194 is an **outlier**.

LINKING QUESTION

What are the principles of effective sampling in biological research? Twenty-five students tell you about all students only if they were chosen fairly (at random, not just the basketball team), if there are enough of them, and if each was measured the same way. A box plot of a poor sample is a neat picture of the wrong thing.

PREDICT

In a box plot, what fraction of the data lie inside the box?

- (A) One quarter
- (B) One half
- (C) All of it, except the outliers

Decide before you look. Answer on page 54.

PREDICT

A student is 194 cm tall. Q_3 is 172 cm and the IQR is 12 cm. Is 194 an outlier?

- (A) No, because only values below the lower quartile can be outliers
- (B) No, because 194 is less than twice the IQR above Q_3
- (C) Yes, because 194 is above $172 + 1.5 \times 12 = 190$

Decide before you look. Answer on page 54.

13

Two genes *at once*

D3.2.16 HL

D3.2.17 HL

SEGREGATION, INDEPENDENT ASSORTMENT AND DIHYBRID CROSSES

A **dihybrid cross** follows two genes. The question is whether the alleles of the two genes travel together or separately. Take Mendel's two seed genes: colour (Y yellow, y green) and shape (R round, r wrinkled).

First, one gene. In meiosis I the two chromosomes of a homologous pair go to opposite poles, so the two alleles of a gene end up in different gametes: each gamete gets one. That is **segregation**.

Now two genes on different chromosomes. In meiosis I each pair lines up on the equator and faces the poles one way or the other, independently of every other pair. A YyRr parent can send Y with R and y with r, or Y with r and y with R. Both are equally likely, so it makes four kinds of gamete, YR, Yr, yR and yr, in equal numbers. This is **independent assortment**.

Cross two YyRr plants. Each makes four kinds of gamete, so a 4 × 4 Punnett grid gives 16 equally likely combinations. Count the phenotypes: 9 yellow round, 3 yellow wrinkled, 3 green round and 1 green wrinkled. That is **9 : 3 : 3 : 1**. It is the product of two 3 : 1 ratios: (3 yellow : 1 green) × (3 round : 1 wrinkled). Cross a double heterozygote YyRr with a double recessive yyrr and the recessive parent adds only yr, so the offspring show the heterozygote's four gametes: **1 : 1 : 1 : 1**.

KEY TERMS

dihybrid cross

A cross that follows two genes at once.

segregation

The separation of the two alleles of a gene into different gametes, because homologous chromosomes go to opposite poles in meiosis I.

independent assortment

The random and independent behaviour of different pairs of chromosomes in meiosis I, so alleles of genes on different chromosomes are inherited independently.

Two genes, two pairs of chromosomes

YyRr × YyRr				
	YR	Yr	yR	yr
YR	 YYRR	 YYRr	 YyRR	 YyRr
Yr	 YYRr	 YYrr	 YyRr	 Yyrr
yR	 YyRR	 YyRr	 yyRR	 yyRr
yr	 YyRr	 Yyrr	 yyRr	 yyrr
9 : 3 : 3 : 1				

 9 yellow, round
 3 yellow, wrinkled
 3 green, round
 1 green, wrinkled

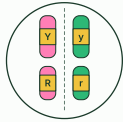
STEP BY STEP

5 STEPS

1

Two genes, two pairs of chromosomes

chromosomes line up in pairs



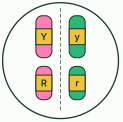
Each pair lines up at the equator. Which partner faces which pole is chance.

In meiosis I each pair of **homologous chromosomes** lines up at the equator, and the partners go to opposite poles.

2

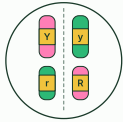
Two genes, two pairs of chromosomes

one way



gametes: YR and yr

or the other way



gametes: Yr and yR

Both ways are equally likely: four kinds of gamete, in equal numbers.

The pairs line up either way round, independently. Two equally likely ways give **four kinds of gamete**: YR, yr, Yr and yR.

3

Two genes, two pairs of chromosomes

YyRr × YyRr

	YR	Yr	yR	yr
YR				
Yr				
yR				
yr				

A YyRr × YyRr cross: put each parent's four kinds of gamete along the sides of a 4 × 4 grid.

4

Two genes, two pairs of chromosomes

YyRr × YyRr

	YR	Yr	yR	yr
YR	YYRR	YYRr	YyRR	YyRr
Yr	YYRr	YYrr	YyRr	Yyrr
yR	YyRR	YyRr	yyRR	yyRr
yr	YyRr	Yyrr	yyRr	yyrr

Fill in the **16 genotypes**, each equally likely.

5

Two genes, two pairs of chromosomes

YyRr × YyRr

	YR	Yr	yR	yr
YR	YYRR	YYRr	YyRR	YyRr
Yr	YYRr	YYrr	YyRr	Yyrr
yR	YyRR	YyRr	yyRR	yyRr
yr	YyRr	Yyrr	yyRr	yyrr

9 : 3 : 3 : 1

9 yellow, round
3 yellow, wrinkled
3 green, round
1 green, wrinkled

Read off the phenotypes: **9 : 3 : 3 : 1**.

ANIMATION · WATCH IT HAPPEN: TWO GENES LINE UP INDEPENDENTLY

9 MOMENTS

1 CELL 1 OF 2

parent YyRr

YR Yr yR yr

from its mother from its father gametes made, by type

2 CELL 1 OF 2

parent YyRr

YR Yr yR yr

from its mother from its father gametes made, by type

One cell of an **YyRr** parent, its DNA already copied. Gene Y is on one pair of chromosomes and gene R on another: capitals from its mother (pink), lower case from its father (green).

Each chromosome pairs with its homologue, so the two alleles of each gene lie side by side.

3 CELL 1 OF 2

parent YyRr

YR Yr yR yr

from its mother from its father gametes made, by type

4 CELL 1 OF 2

parent YyRr

YR Yr yR yr

from its mother from its father gametes made, by type

The pairs line up. In this cell both pink chromosomes face the same end, so Y will go with R, and y with r.

Segregation: in meiosis I the alleles of each gene separate, Y from y and R from r.

5 CELL 1 OF 2

parent YyRr

YR Yr yR yr

from its mother from its father gametes made, by type

6 CELL 1 OF 2

parent YyRr

YR Yr yR yr

from its mother from its father gametes made, by type

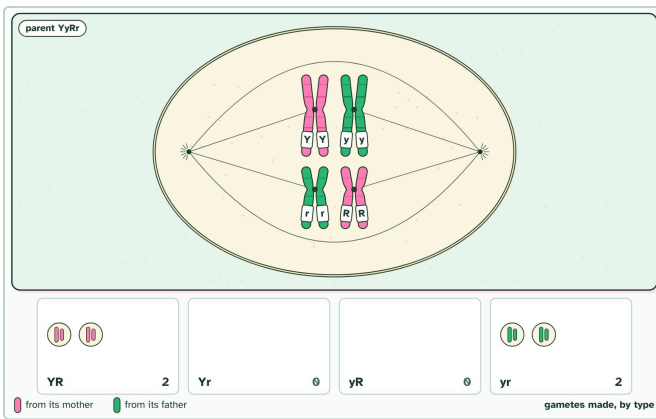
Meiosis II separates the sister chromatids, which carry the same alleles.

This cell makes two kinds of gamete: YR and yr.

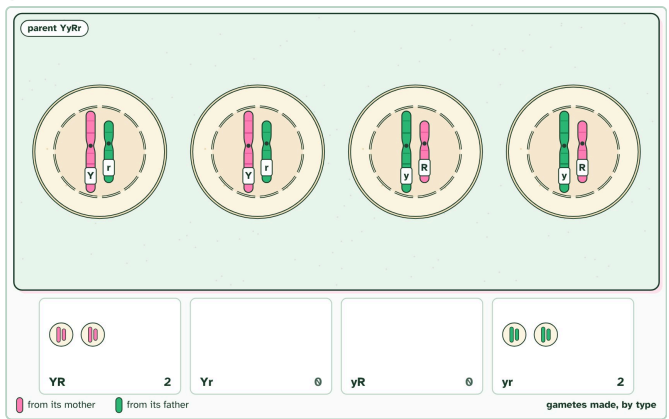
7

CELL 2 OF 2 8

CELL 2 OF 2



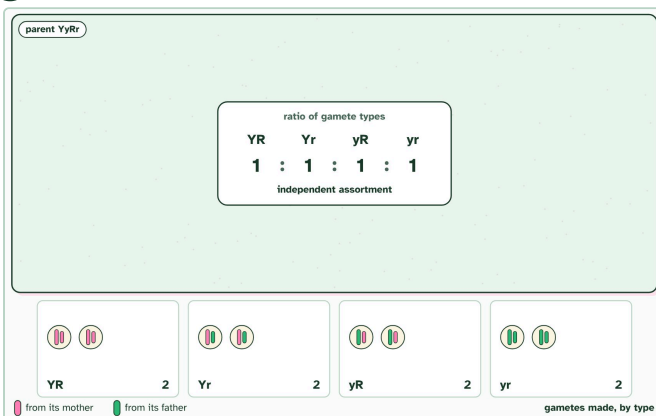
In another cell the pairs line up the other way round: now Y goes with r, and y with R.



That cell makes Yr and yR gametes.

9

8 GAMETES MADE



Both line-ups are equally likely, so the four types are made in equal numbers, **1:1:1:1**. This is **independent assortment**.

Watch it move, and change it as it runs: biologybybradford.com/guides/ib-biology-d3-2-inheritance#dihybrid

P generation

female parent
YYRR
 yellow, round

x

male parent
yyrr
 green, wrinkled

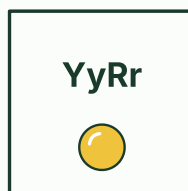
F1 offspring

male gametes, in pollen

yr

female gametes

YR

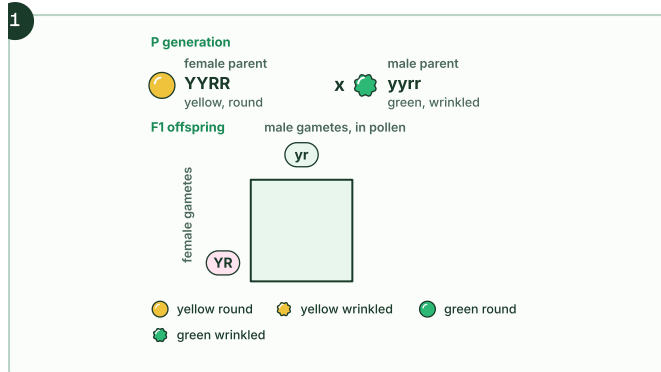


yellow round
 yellow wrinkled
 green round
 green wrinkled

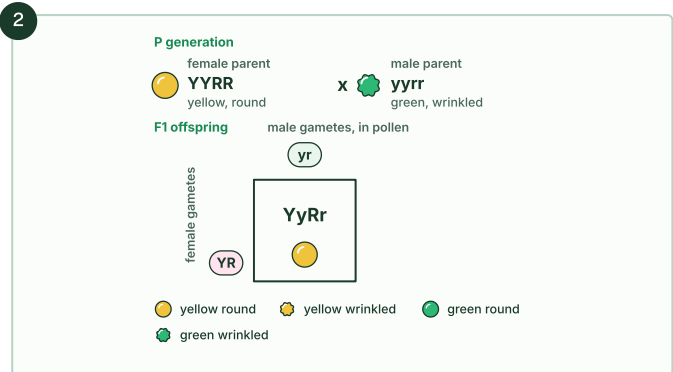
Choose two parents for the seed colour and shape genes. Predict the ratio, then see the 16-box grid fill and breed a sample.

STEP BY STEP

2 STEPS



Each parent makes gametes by meiosis, one allele of each gene in each. The female parent's go down the side of the **Punnett grid**, the male parent's along the top.



Each cell joins one gamete from each parent: a possible offspring, every cell as likely as the next. The F1: genotypes all YyRr; phenotypes all yellow round.

STEPS

(1) Write both parents' genotypes. (2) Write each parent's gametes: one allele of each gene, so a YyRr parent gives YR, Yr, yR and yr. (3) Put one parent's gametes along the top and the other's down the side, and fill in the grid. (4) Count the phenotypes, remembering that capitals show.

NATURE OF SCIENCE

These ratios are based on what has been called Mendel's second law. It only applies if the genes are on different chromosomes, or far enough apart on one chromosome that recombination reaches 50%. Chapter 14 shows genes that break it. There are exceptions to all biological "laws" under some conditions.

PREDICT

A plant YyRr is crossed with a plant yyrr. What proportion of the offspring are yellow and round?

- (A) One half
(B) One quarter
(C) Nine sixteenths

Decide before you look. Answer on page 54.

PREDICT

What must be true for the 9 : 3 : 3 : 1 ratio to appear?

- (A) Both genes must be dominant over each other in every cross
(B) The two genes are on different chromosomes or far apart
(C) Both parents must be homozygous for each gene

Decide before you look. Answer on page 54.

14

Genes that *travel together*

D3.2.18 HL

D3.2.19 HL

D3.2.20 HL

GENE LOCI, LINKAGE AND RECOMBINANTS

Independent assortment works only if the genes are on different chromosomes. A chromosome carries hundreds or thousands of genes, so most genes share a chromosome with others. Genes on the same chromosome are **linked**: they travel together in meiosis because they sit on the same piece of DNA.

A gene database lists each human gene with its locus (the chromosome and the position on it) and the polypeptide it codes for, so you can look pairs up. Try the genes for the beta chain of haemoglobin (*HBB*) and the alpha chain (*HBA1* and *HBA2*): they sit on different chromosomes, so they assort independently. Then look at the genes next to *HBB*, such as *HBD* for the delta chain: they are close neighbours on the same chromosome, so they are linked. Your teacher will name a database.

Linked alleles are shown with a vertical line for each homologous chromosome and the alleles beside it: AB over ab means A and B are on one chromosome and a and b on its partner. With no crossing over, only two kinds of gamete form, AB and ab: the combinations that were already on the chromosomes, called **parental** types.

But chromosomes do swap pieces. In prophase I, homologous chromosomes pair up and non-sister chromatids can cross over. If a crossover falls between the two genes, some gametes get Ab or aB. A gamete, a genotype or a phenotype with a combination of alleles that differs from the combinations on the parent's chromosomes is a **recombinant**. The further apart two genes are, the more often a crossover falls between them, so the more recombinants. Genes far enough apart to give 50% recombinants behave as if unlinked.

The **test cross** shows this. Cross an AaBb individual with an aabb one. The aabb parent adds only *ab*, so each offspring's phenotype shows the gamete it got from the heterozygote. Unlinked genes give 1:1:1:1, half of them recombinants. Linked genes give two large parental classes and two small recombinant classes. The **recombinant frequency** is the recombinants divided by all the offspring. You can find recombinants in gametes, in the genotypes of offspring and in their phenotypes.

Try it yourself in the dry lab: choose unlinked genes or genes linked at different distances, set up a test cross and count the offspring. Or try the linked or not? mystery.

KEY TERMS

linked

Genes with their loci on the same chromosome, whose alleles tend to be inherited together.

recombinant

A gamete, genotype or phenotype with a combination of alleles that the parent chromosomes did not have, made by crossing over or by independent assortment.

test cross

A cross between an individual heterozygous for both genes and one homozygous recessive for both. The offspring's phenotypes show which gametes the heterozygote made.

Genes on one chromosome travel together

200 offspring of AaBb × aabb (made-up numbers)

number of offspring



recombinants: $16 + 14 = 30$ of 200, which is 15%

STEP BY STEP

4 STEPS

1 Genes on one chromosome travel together

two different chromosome pairs

meiosis

four kinds, in equal numbers

Unlinked genes sit on different chromosome pairs, so all four kinds of gamete are equally likely.

2 Genes on one chromosome travel together

the same chromosome pair

meiosis, no crossing over

only two kinds of gamete

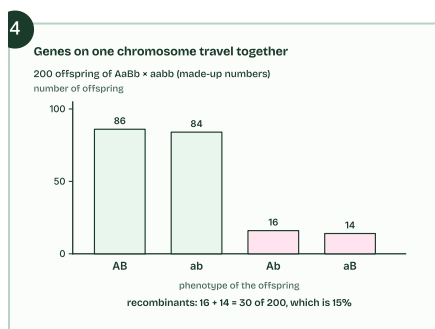
Linked genes sit on the same chromosome. With no crossing over, only the two **parental** gametes, AB and ab, are made.

3 Genes on one chromosome travel together

crossing over

many few few many

Crossing over between the genes swaps the ends of two chromatids and makes the **recombinant** gametes Ab and aB. The other two chromatids are unchanged, so recombinants are few.



A **test cross** (AaBb × aabb) shows it: mostly parental types and a few recombinants. Recombinant frequency = $30 \div 200 = 15\%$.

WATCH OUT

Linked genes are not "always inherited together": crossing over separates them sometimes. And a recombinant is not a mutant. No allele has changed; only the combination of alleles is new.

PREDICT

A test cross $AaBb \times aabb$ gives 48 AB, 47 ab, 3 Ab and 2 aB. What does this suggest?

- ☐ (A) The genes are unlinked, so all four classes should be equal
- ☐ (B) The genes are on different chromosomes, with recombinants common
- ☐ (C) The genes are linked, and crossovers are rare between them

Decide before you look. Answer on page 54.

PREDICT

Two genes lie far apart at opposite ends of a long chromosome. What will a test cross look like?

- ☐ (A) Almost all parental types, with very few recombinants
- ☐ (B) About equal numbers of all four types, like unlinked genes
- ☐ (C) Only the two recombinant types, because crossing over always happens

Decide before you look. Answer on page 54.

15

D3.2.21 HL THE CHI-SQUARED TEST

Is it *chance*? The chi-squared test

Real offspring never match the expected ratio exactly. From 160 plants from a 9 : 3 : 3 : 1 cross you would not expect exactly 90, 30, 30 and 10, even when the genes are unlinked, because chance decides which gametes meet. So when is a difference too big to blame on chance? A **chi-squared test** answers that.

Begin with two hypotheses. The **null hypothesis** says there is no real difference between observed and expected: the genes are unlinked, and chance explains the scatter. The alternative hypothesis says there is a real difference, for example that the genes are linked.

The test gives $\chi^2 = \frac{(O - E)^2}{E}$ over every class, where O is observed and E is expected. The bigger the differences, the bigger χ^2 . Compare it with a critical value from a table, chosen by the degrees of freedom (the number of classes minus 1, so 3 for four phenotype classes) and the significance level, $p = 0.05$. If χ^2 is bigger than the critical value, the difference is statistically significant: a difference this large would arise by chance alone less than 5% of the time, so you reject the null hypothesis. If χ^2 is smaller, chance could explain the difference and you do not reject it.

The dry lab does the arithmetic with you: breed a brood and test it, then decide whether to reject the null hypothesis.

KEY TERMS

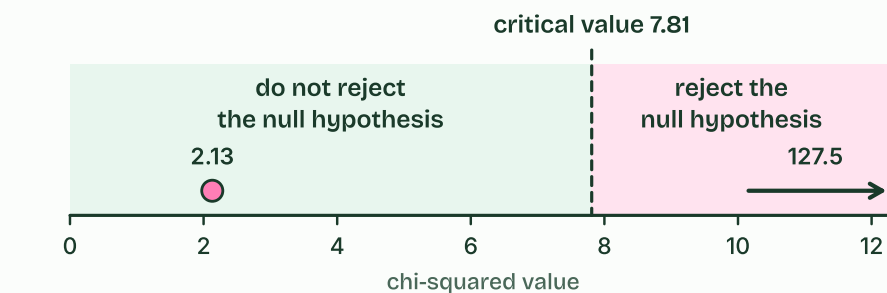
chi-squared test

A statistical test that compares observed counts with the counts expected if a hypothesis is true, to judge whether the difference could be due to chance alone.

null hypothesis

The hypothesis that there is no real difference between the observed and expected results, so that any difference is due to chance.

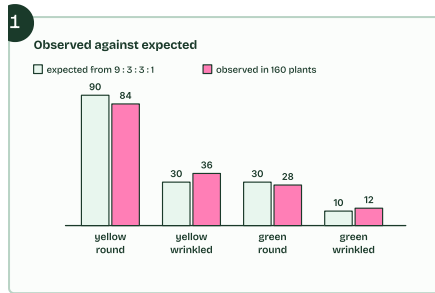
Observed against expected



df = 4 classes - 1 = 3, at $p = 0.05$

STEP BY STEP

4 STEPS

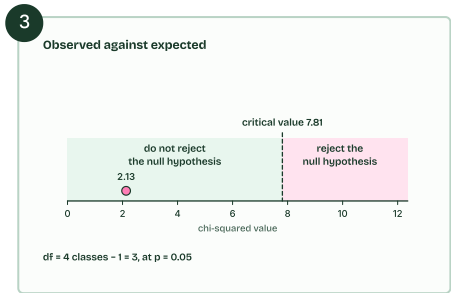


160 F2 plants: the **expected** counts from 9 : 3 : 3 : 1 (90, 30, 30 and 10) against **observed** counts (made up).

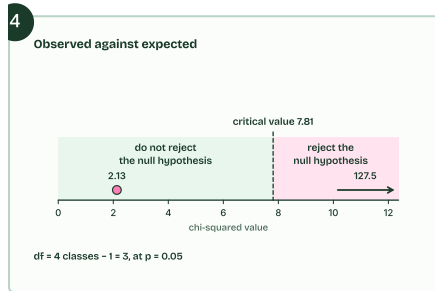
2 Observed against expected

class	O	E	$(O - E)^2 / E$
yellow round	84	90	0.40
yellow wrinkled	36	30	1.20
green round	28	30	0.13
green wrinkled	12	10	0.40
sum = chi-squared			2.13

χ^2 is the sum of $(O - E)^2 \div E$ over the four classes: **2.13**.



With 3 degrees of freedom and $p = 0.05$ the **critical value** is 7.81. 2.13 is below it, so you do not reject the null hypothesis: chance can explain the difference.



A test cross of linked genes (made-up counts 112, 21, 18 and 109 from 260 offspring) gives $\chi^2 = 127.5$, far past 7.81. Reject the null: the genes are probably linked.

REMEMBER IT

The five steps of a chi-squared test

H Hungry	E Elves	C Cook	D Delicious	C Cakes
Hypotheses state the null and the alternative	Expected work out the expected counts	Calculate $\chi^2 = \text{sum of } (O - E)^2 \div E$	Degrees of freedom number of classes minus 1	Compare with the critical value at $p = 0.05$

CRITICAL VALUES AT $P = 0.05$

1 degree of freedom: 3.84. 2: 5.99. 3: 7.81. 4: 9.49.

NATURE OF SCIENCE

Statistical testing often uses a sample to represent a population. Here the sample is the F2 generation: 160 plants standing for every plant this cross could ever produce. In many experiments the sample is the set of repeated measurements. A bigger sample gives a more reliable test, so geneticists breed many offspring and repeat crosses. That is effective sampling, the linking question of this subtopic.

WATCH OUT

A result that is not significant does not prove the null hypothesis: it means these data are not enough to reject it. And a significant result says the data differ from the expected ratio, not why. Linkage is the explanation to test next. The 5% level is a convention: about 1 test in 20 on unlinked genes would look significant by chance alone.

PREDICT

A chi-squared value of 2.1 with 3 degrees of freedom is compared with a critical value of 7.81. What do you conclude?

- ☐ (A) Reject the null hypothesis, because the result is significant
- ☐ (B) Do not reject the null: chance could explain it
- ☐ (C) Accept the alternative, because the observed numbers differ

Decide before you look. Answer on page 55.

PREDICT

You keep every percentage the same but double the number of offspring counted. What happens to chi-squared?

- ☐ (A) It stays the same, because the proportions in each class are the same
- ☐ (B) It halves, because each class has more offspring to spread the error over
- ☐ (C) It doubles: a bigger sample makes the difference more convincing

Decide before you look. Answer on page 55.

Key terms

Every term this guide defines, A to Z, with the chapter it is in. Cover the right-hand side and test yourself.

ABO blood group 07

The system of human blood groups A, B, AB and O, controlled by one gene with three alleles.

alleles 03

Alternative forms of the same gene, found at the same place on a chromosome. They differ in their base sequences.

autosomes 01

Any chromosome that is not a sex chromosome. Autosomes come in matching pairs.

box-and-whisker plot 12

A graph that shows the minimum, first quartile, median, third quartile and maximum of a data set as a box with two whiskers, and any outliers as separate points.

carrier 06

Someone who has one copy of a recessive allele for a condition, is not affected by it, and can pass the allele on.

chi-squared test 15

A statistical test that compares observed counts with the counts expected if a hypothesis is true, to judge whether the difference could be due to chance alone.

codominance 08

A pattern of inheritance in which both alleles of a heterozygote are fully expressed, giving a dual phenotype.

continuous variation 11

Variation in which individuals can have any value across a range, with no separate categories, such as height or skin colour.

deductive reasoning 10

Reasoning from a general rule to a conclusion about a particular case. If the rule and the facts are true, the conclusion is certain.

dihybrid cross 13

A cross that follows two genes at once.

diploid 01

A cell with two sets of chromosomes, one from each parent (2n). It carries two copies of each autosomal gene.

discrete variation 11

Variation in which individuals fall into separate categories with nothing in between, such as blood group.

dominant allele 04

An allele whose effect shows in the phenotype of a heterozygote as well as of a homozygote.

gametes 01

A sex cell, such as a sperm, an egg or a pollen grain, with one set of chromosomes.

gene 03

A length of DNA at one place (locus) on a chromosome, with the instructions for making a polypeptide.

gene expression 05

The use of the information in a gene to make a product, so that the gene affects the phenotype.

gene pool 07

All the alleles of all the genes in a population.

genotype 03

The combination of alleles an organism has, usually for one gene.

haemophilia 09

An X-linked recessive disorder in which blood clots slowly or not at all, because a protein needed for clotting is missing or does not work.

haploid 01

A cell with one set of chromosomes (n), such as a gamete.

heterozygous 03

Having two different alleles of a gene.

homozygous 03

Having two identical alleles of a gene.

incomplete dominance 08

A pattern of inheritance in which the heterozygote has a phenotype in between those of the two homozygotes.

independent assortment 13

The random and independent behaviour of different pairs of chromosomes in meiosis I, so alleles of genes on different chromosomes are inherited independently.

inductive reasoning 10

Reasoning from observations of some cases to a general conclusion or theory. The conclusion is probable, not certain.

interquartile range 12

Interquartile range: Q_3 minus Q_1 , the spread of the middle half of the data.

linked 14

Genes with their loci on the same chromosome, whose alleles tend to be inherited together.

meiosis 01

A type of cell division that halves the chromosome number. It makes the gametes of an organism that reproduces sexually.

null hypothesis 15

The hypothesis that there is no real difference between the observed and expected results, so that any difference is due to chance.

outlier 12

A data point more than 1.5 times the IQR above the third quartile or below the first quartile.

pedigree chart 10

A chart of the members of a family over several generations, showing who is affected by a genetic condition.

phenotype 03

The observable traits of an organism, which result from its genotype and the environment.

phenotypic plasticity 05

The ability of one genotype to produce different phenotypes, by changing which genes are expressed, in response to the environment the organism experiences.

phenylketonuria 06

A recessive genetic condition in which the body cannot convert phenylalanine to tyrosine, because the enzyme that does this is not working.

pollen 02

The tiny grains made in the anthers of a flower. Each pollen grain contains male gametes.

pollination 02

The transfer of pollen from an anther to a stigma.

polygenic 11

Controlled by several genes, each adding a small effect.

Punnett grid 02

A grid that shows every way the gametes of two parents can combine, so you can read off the genotypes of the offspring and how likely each is.

quartiles 12

The values that split ordered data into four equal groups. A quarter of the data lie below the first quartile (Q_1) and a quarter above the third (Q_3).

recessive allele 04

An allele whose effect shows only when there is no dominant allele, so the organism is homozygous for it.

recombinant 14

A gamete, genotype or phenotype with a combination of alleles that the parent chromosomes did not have, made by crossing over or by independent assortment.

segregation 13

The separation of the two alleles of a gene into different gametes, because homologous chromosomes go to opposite poles in meiosis I.

self-fertilisation 02

Fertilisation of a plant's own ovules by its own pollen.

sex-linked 09

A gene on a sex chromosome, so its pattern of inheritance differs between males and females.

single-nucleotide polymorphism 07

Single-nucleotide polymorphism: a position in the genome where individuals differ by one base, and where each version is common in the population.

test cross 14

A cross between an individual heterozygous for both genes and one homozygous recessive for both. The offspring's phenotypes show which gametes the heterozygote made.

true-breeding 02

Plants or animals that always produce offspring like themselves when bred with their own kind, because both alleles of the gene are the same.

zygote 01

The diploid cell made when two gametes fuse. It divides and grows into a new organism.

Dry lab: *Fly crosses: linked or not?*

Cross flies that carry two invented genes, count each brood and use a chi-squared test to decide whether the genes assort independently or travel together. Then try the mystery pairs. This bench is for higher level.

F1 parent
heterozygous for both genes

W straight, w curled wings; R red, r plum eyes

Test-cross parent
double recessive

invented genes, not real fly data

×

Gametes from the F1 parent

W R

parental · 40 %

w r

recombinant · 10 %

w R

recombinant · 10 %

W r

parental · 40 %

200 offspring a brood

**straight wings
red eyes**

**straight wings
plum eyes**

**curled wings
red eyes**

**curled wings
plum eyes**

The settings



Run it online: change the variables, run the trials, and the table and graph fill in as you go. biologybybradford.com/guides/ib-biology-d3-2-inheritance#lab

Your question

Do two genes on the same chromosome sort independently, as genes on different chromosomes do? Start with one pair of genes and one cross, work out the ratio you expect if they are unlinked, then see what a real brood gives you.

What you can change

Whether the two genes sit on different chromosomes or are linked at 40, 20 or 5 percent recombination, the cross (a test cross or F1 by F1), and the size of the brood (100, 200, 500 or 1000). The genes are invented and are not real fruit fly data. Change one thing at a time.

What to record

For each brood: the settings, the count of each of the four phenotypes, the percentage of recombinants, the expected counts, your chi-squared value, the degrees of freedom, and your decision to reject or not reject the null hypothesis. Repeat each set-up at least three times: broods scatter, as real ones do. The coach under the bench helps you plan, process and evaluate.

Find out

- **What do unlinked genes give?** Set the two genes on different chromosomes, make a test cross and breed 200 offspring. Write down the ratio you expect before you breed. How close is the brood to it, and how far apart are the four classes?
- **What does linkage do to the ratio?** Repeat with the genes linked at 40, then 20, then 5 percent recombination. Which two classes grow, which two shrink, and which classes are the recombinants?
- **Does the size of the brood matter?** Use 100, then 500, then 1000 offspring for the same linked pair. At what size does the chi-squared test start to reject the unlinked expectation every time?
- **Can you tell by looking?** Open the chi-squared working for one brood. Say which hypothesis you are testing, work out the expected counts, then let the bench check your value and the critical value for 3 degrees of freedom.
- **Linked or not?** Switch to the mystery. You get hidden pairs of genes. Breed as many broods as you need, give your verdict, and see how often you are right.

Record

- For every brood: the two genes, how they are placed (different chromosomes or linked, and how far), the cross and the number of offspring.
- The count of each of the four phenotypes, the percentage that are recombinants, and the expected count of each class if the genes are unlinked.
- Your chi-squared value, the degrees of freedom, the critical value at $p = 0.05$, and your decision to reject or not reject the null hypothesis.
- Change one setting at a time. Repeat each set-up at least three times: no two broods match, because chance decides which gametes meet.
- Use the live graph to compare observed and expected bars, or download the CSV and plot your own. The genes here are invented, so no result is a fact about real flies.

QUESTIONS ON THE LAB

QUESTION 1

You make a test cross with two genes on different chromosomes and breed 200 offspring. What do you expect for the four phenotypes?

- (A) About 150 with both dominant traits and about 50 with both recessive traits, and almost none of the mixed kinds
- (B) About 50 of each of the four, though no brood will hit those numbers exactly
- (C) About 112 and 12 of two kinds and about 38 of each of the others, following the 9 : 3 : 3 : 1 ratio

Answer on page 55.

QUESTION 2

Now the two genes are linked, with 20 percent recombination, and you breed 200 offspring from a test cross. Which result fits best?

- (A) About 80 and 80 of the two parental types, and about 20 and 20 of the two recombinant types
- (B) About 50 of each type, because the genes are still sorted into gametes at random, linked or not
- (C) About 100 and 100 of the two parental types, and none of the recombinant types, because the genes stay on one chromosome

Answer on page 55.

QUESTION 3

A brood from a test cross gives a chi-squared value above the critical value for the unlinked expectation. What should you conclude?

- (A) The genes are definitely linked, because a value that high can never happen by chance alone, whatever the cross
- (B) The experiment has failed, because a correct brood should always match the expected ratio exactly, as the grid says
- (C) Reject the null hypothesis: chance alone is unlikely to explain it, so the genes are probably linked

Answer on page 55.

WRITE IT UP AS AN INVESTIGATION

Your results from this dry lab can be written up the way an IB scientific investigation is: four criteria, each worth 6 marks. Tick each line as your write-up does it. The coach on the page checks these as you go; it never gives a mark.

1 Research design

A focused question set in its biology; your choices of what to measure, how, over what range and how often, explained rather than listed; what you kept the same and how; safety, ethics and the environment; a method someone else could follow and repeat.

- ☐ My question names what I changed.
- ☐ It gives the range I used, with units.
- ☐ It names what I measured and how.
- ☐ It names the organism or system.
- ☐ My background explains, with the biology, why the two should be linked.
- ☐ I cite my sources, and each reference can be traced.
- ☐ If I give a hypothesis, it predicts and explains.
- ☐ Every variable has its How, Why and Units.
- ☐ For each thing kept the same, I say how I kept it so.
- ☐ I list at least three things I kept the same.
- ☐ I name the apparatus that set what I changed, and the instrument that measured.
- ☐ I test enough values.
- ☐ My values spread wide enough.
- ☐ I run at least three trials at every value.
- ☐ I run a control.
- ☐ I explain why these values, this many and this often.
- ☐ Someone else could repeat my method.
- ☐ I deal with safety, ethics and the environment.

3 Conclusion

Your question answered from the processed data, with numbers quoted and the spread taken into account; the biology that explains the pattern; a comparison with published work; no claim bigger than the data.

- ☐ I answer my question in a sentence, naming both variables.
- ☐ I quote my processed numbers.
- ☐ I say how sure the spread lets me be.
- ☐ I say whether my hypothesis is supported.
- ☐ I explain the pattern with biology.
- ☐ I compare my result with published work, and cite it.
- ☐ I claim no more than my data show.

2 Data analysis

Raw data recorded clearly, with titles, units, uncertainties and the same decimal places down a column; what you saw as well as what you measured; processing that suits the question, shown with a worked example; the spread shown; a graph a reader can trust.

- ☐ Each table has a numbered title that says what it shows.
- ☐ Every heading has its quantity and its unit.
- ☐ Every raw data heading has its uncertainty.
- ☐ Each uncertainty is realistic for the instrument.
- ☐ Each uncertainty has the same decimal places as its readings.
- ☐ I record what I saw as well as what I measured.
- ☐ I show one worked example of my processing, and it is right.
- ☐ I say why I left out any trial.
- ☐ My error bars show a spread my repeats can support.
- ☐ My graph has a numbered title and labelled axes with units.
- ☐ I say what the error bars show.
- ☐ A straight line, if I draw one, has its R^2 .
- ☐ A statistical test, if I run one, has its hypotheses and suits the data.

4 Evaluation

Weaknesses and limitations of this method (not slips in carrying it out), with evidence from your own data, the effect each had on the results, which mattered most and why, and a realistic improvement for each.

- ☐ I give at least three weaknesses or limitations.
- ☐ Every row is filled in.
- ☐ No slips in carrying it out: weaknesses of the method itself.
- ☐ Each improvement is specific: which instrument, how many, what changes.
- ☐ Each effect says what it did to my data.
- ☐ At least two rows point to evidence in my own data.
- ☐ At least one limitation: what this experiment cannot tell me.
- ☐ I say which mattered most, and why.
- ☐ I deal with what stands out in my data.
- ☐ My improvements improve this investigation, not a new one.

Variables

Variable	How?	Why?	Units
Independent: position of the two genes (where their loci sit)			
Dependent: percentage of recombinant offspring			
Kept the same: the cross: F1 heterozygote x double recessive (a test cross)			
Kept the same: number of offspring counted in each brood			
Kept the same: the two genes studied (wing shape and eye colour)			
Kept the same: how the alleles sit in the F1 parent (both dominant alleles from one pure-breeding parent)			
Kept the same: temperature the flies are raised at			
Kept the same: food medium and vial size			
Kept the same: when the offspring are counted (after all have emerged)			

Evaluation

Weakness or limitation	Which?	Evidence in my data	Effect on my results	Improvement

Data generated with the fly cross dry lab, biologybybradford.com (a simulation of invented genes in an invented fly; not real fruit fly data). These checks catch what is missing. They cannot judge how good it is: that is what your teacher does.

**Check yourself**

The check for this guide is online: a fresh paper of past-paper style questions from the exam bank on D3.2 each time, marked as you go, and saved to your Exam practice progress when you are signed in. Scan the code, or type the link.

biologybybradford.com/exam-maker/sit?topics=D3.2&level=HL&paper=quiz&marks=15&src=ep

Answers

PREDICTIONS IN THE CHAPTERS

- 01** A human body cell has 46 chromosomes. How many chromosomes are in a sperm, and how many in the zygote it helps to form?
- B 23 in the sperm and 46 in the zygote**
- 23 and 46. Meiosis halves the number, so a sperm is haploid with one chromosome from each of the 23 pairs. The zygote gets 23 from the sperm and 23 from the egg, so it is diploid again. If gametes were made by an ordinary division, the number would double every generation.
-
- 02** A student wants to cross a purple-flowered pea plant with a white-flowered one, but forgets to remove the anthers from the white plant. What is likely to happen?
- A Some seeds will come from self-fertilisation, so not all will be from the cross**
- Some seeds will come from self-fertilisation. A pea flower makes both pollen and ovules and normally pollinates itself, so unless the anthers are removed early, its own pollen can fertilise some of the ovules. The offspring would be a mixture of crossed and self-fertilised seeds, and the results would be hard to read.
-
- 03** Two plants have the same genotype for every gene, but one grows much taller because it was given more water and fertiliser. What does this show?
- B The environment affects the phenotype as well as the genotype**
- The environment affects the phenotype as well as the genotype. The two plants have the same alleles, so the difference in height comes from the conditions they grew in. Fertiliser does not change the alleles in the plant's cells.
-
- 03** A pea plant has the genotype Yy. Which statement is correct?
- B It is heterozygous, because its two alleles of the gene differ**
- It is heterozygous. Both alleles belong to the same gene, the seed colour gene, but they are different versions of it. Homozygous means the two alleles are the same. A plant does not have two different genes for one trait; it has two alleles of one gene.
-
- 04** Two yellow-seeded pea plants are crossed, and some of their offspring have green seeds. What must be true of both parents?
- B Both are heterozygous Yy: each carries a hidden y allele**
- Both are Yy. A green seed is yy, so each parent must have given it a y. A parent that shows the dominant trait yet carries y must be heterozygous. A YY parent has no y to give, so one YY parent could never produce a green seed.
-
- 04** Two Yy plants are crossed. What fraction of their offspring have green seeds?
- C One quarter**
- One quarter. Each parent makes Y and y gametes in equal numbers. A green seed needs a y from both parents, which has a chance of $1/2 \times 1/2 = 1/4$. Use the tool below to check, and then breed 30 offspring to see how a real sample scatters around the prediction.
-
- 05** Two clones grow differently, one in sun and one in shade. What explains the difference?
- B The light changed how their genes were expressed**
- The light changed how their genes were expressed. Clones start with identical alleles, and nothing in this story changes them. The environment changes which genes are switched on and how strongly, and that changes the phenotype.
-

05 Which of these is an example of phenotypic plasticity?

C A person's skin tans in strong sun and fades in winter

The tan. The same person, with the same alleles, develops different skin colour in different conditions, and it can reverse. A blood group is inherited and does not change in life. A population darkening over generations is evolution by natural selection, which changes allele frequencies.

06 Two parents do not have PKU, but their first child does. What is the chance that their second child has PKU?

C 1 in 4, because both parents must be carriers

1 in 4. The child is nn , so each unaffected parent must be Nn . Two Nn parents have a 1 in 4 chance of nn for every child. Alleles are not used up: each new baby is made from fresh gametes.

06 A baby with PKU is put on a diet low in phenylalanine. Has its genotype changed?

B No: the genotype is the same, but the phenotype changes

No. The baby still has two faulty alleles. The diet changes the environment, so less phenylalanine builds up, and the phenotype is different. This is the same story as the pea plants: the phenotype comes from genotype and environment together.

07 A man with blood group AB ($I^A I^B$) and a woman with group O (ii) have children. Which groups are possible?

C A and B only

A and B only. The man's gametes carry I^A or I^B . The woman's all carry i . So every child is $I^A i$ (group A) or $I^B i$ (group B). Neither AB nor O can occur, even though the parents are AB and O.

07 How many alleles of the ABO gene does one person have?

B Two, one from each parent

Two. The gene pool has three alleles, but each person is diploid and inherits one from each parent. A person who is homozygous (such as ii) has two copies of the same allele.

08 A red four o'clock plant is crossed with a white one. What colour are the F1 flowers?

C All pink

All pink. Every F1 plant is $C^R C^W$, a heterozygote, and in incomplete dominance the heterozygote is intermediate. Crossing two of these pink plants then gives 1 red : 2 pink : 1 white.

08 What blood group does a person with the genotype $I^A I^B$ have, and why?

B Group AB, because both alleles are expressed

Group AB. I^A and I^B are codominant, so both are expressed and the red blood cells carry both markers. This is a dual phenotype, not an intermediate one.

09 A man with haemophilia ($X^h Y$) and a woman who is not a carrier ($X^H X^H$) have children. What are the children like?

C All daughters are carriers; all sons are unaffected

All daughters are carriers and all sons are unaffected. Every daughter gets her father's X^h and her mother's X^H , so she is a carrier. Every son gets the father's Y and the mother's X^H , so he is unaffected.

09 Why are X-linked recessive conditions more common in males than in females?

C Males have one X, so one recessive allele shows

Males have one X, so one recessive allele shows. A female needs two copies of the allele, one on each X, to be affected. Males also get their X from their mother, not their father, which is the reason fathers do not pass these alleles to sons.

- 10 In a pedigree, two unaffected parents have an affected daughter. Which statement is correct?

C It is recessive and autosomal, so both parents carry it

Recessive and autosomal. Unaffected parents with an affected child must both carry a hidden recessive allele. And an affected daughter with an unaffected father rules out an X-linked recessive condition, because she would need an affected father to get the X allele from him.

- 10 You decide, from several families, that a trait is probably recessive. What kind of reasoning is this?

B Inductive: it generalises from the families seen

Inductive. You move from observed cases to a general idea that may fit more cases. Working out one person's genotype once the pattern is accepted is deductive.

- 11 Which of these shows discrete variation?

B The blood groups of students in a class

The blood groups. Every student is A, B, AB or O, with nothing in between, so the variation falls into separate categories. Height and seed mass can take any value in a range, which is continuous variation.

- 12 In a box plot, what fraction of the data lie inside the box?

B One half

One half. The box runs from the first quartile to the third quartile, and a quarter of the data are below the first quartile and a quarter above the third, so the middle half is in the box.

- 12 A student is 194 cm tall. Q_3 is 172 cm and the IQR is 12 cm. Is 194 an outlier?

C Yes, because 194 is above $172 + 1.5 \times 12 = 190$

Yes. The upper fence is $Q_3 + 1.5 \times IQR = 172 + 18 = 190$, and 194 is beyond it. Values more than $1.5 \times IQR$ above Q_3 or below Q_1 count as outliers.

- 13 A plant YyRr is crossed with a plant yyrr. What proportion of the offspring are yellow and round?

B One quarter

One quarter. The yyrr parent makes only yr gametes, so the offspring show the four gametes of the YyRr parent, in equal numbers: YyRr (yellow round), Yyrr (yellow wrinkled), yyRr (green round) and yyrr (green wrinkled). One of the four is yellow and round.

- 13 What must be true for the 9 : 3 : 3 : 1 ratio to appear?

B The two genes are on different chromosomes or far apart

The two genes must be on different chromosomes, or far enough apart on one chromosome to assort independently. If they are close together they travel as a block, and the ratios change (chapter 14). The parents must in fact be heterozygous for both genes to give this ratio.

- 14 A test cross AaBb × aabb gives 48 AB, 47 ab, 3 Ab and 2 aB. What does this suggest?

C The genes are linked, and crossovers are rare between them

The genes are linked, and crossovers between them are rare. The two parental types, AB and ab, are far commoner than the recombinants Ab and aB. Unlinked genes would give four roughly equal classes.

- 14 Two genes lie far apart at opposite ends of a long chromosome. What will a test cross look like?

B About equal numbers of all four types, like unlinked genes

About equal numbers of all four types. With a long stretch between them, a crossover falls between the genes in about half of meioses, which gives 50% recombinants, the same as for unlinked genes.

- 15** A chi-squared value of 2.1 with 3 degrees of freedom is compared with a critical value of 7.81. What do you conclude?

B Do not reject the null: chance could explain it

Do not reject the null hypothesis. 2.1 is below 7.81, so a difference this size can easily arise by chance when the genes are unlinked. The numbers do differ from the expected ones, but not by more than chance alone would explain.

- 15** You keep every percentage the same but double the number of offspring counted. What happens to chi-squared?

C It doubles: a bigger sample makes the difference more convincing

It doubles. If O and E are both doubled, $(2O - 2E)^2 \div 2E = 2 \times (O - E)^2 \div E$. The same proportional difference in a bigger sample is stronger evidence, which is why larger samples matter.

QUESTIONS ON THE DRY LAB

- Q1** You make a test cross with two genes on different chromosomes and breed 200 offspring. What do you expect for the four phenotypes?

B About 50 of each of the four, though no brood will hit those numbers exactly

About 50 of each. A test cross crosses a double heterozygote with a double recessive, and unlinked genes assort independently, so the four gamete types are equally likely and the offspring come out 1:1:1:1. The 9:3:3:1 ratio belongs to a cross between two double heterozygotes. Expect scatter around 50 each, which is why we test.

- Q2** Now the two genes are linked, with 20 percent recombination, and you breed 200 offspring from a test cross. Which result fits best?

A About 80 and 80 of the two parental types, and about 20 and 20 of the two recombinant types

80, 80, 20 and 20. Twenty percent of 200 is 40 recombinants, shared equally between the two recombinant types. The parental combinations stay together most of the time, so they make up the other 160. If crossing over never happened, you would get only the two parental types, but crossing over between the loci makes some recombinants, so a few of each of the other two appear.

- Q3** A brood from a test cross gives a chi-squared value above the critical value for the unlinked expectation. What should you conclude?

C Reject the null hypothesis: chance alone is unlikely to explain it, so the genes are probably linked

Reject the null hypothesis. If the genes were unlinked, a value that large would come up less than 5 times in 100, so the difference is probably not chance. That is evidence for linkage, not proof: about 1 brood in 20 from unlinked genes will be rejected, and the bench lets you see that happen by breeding many broods.

How this was made. The course structure, the activities and the teaching choices come from my own classroom, my presentations and my years of teaching IB and AP Biology. I use Claude, an AI, to help write and build the interactive guides, lesson plans, slides and practice questions, and to draw the diagrams and code the interactives. I direct the work and review it against the IB and AP specifications before it goes live.

Written and drawn for Biology by Bradford, biologybybradford.com. Free to print and share with a class.