

Inheritance

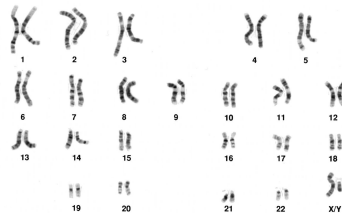
A-Level Biology ■ Topic 16

A-Level Biology

TT	Tt
Tt	tt

What we will cover

- 1 Meiosis & variation
- 2 The language of genetics
- 3 Genetic crosses
- 4 Sex linkage
- 5 Genes to phenotype
- 6 Gene control



Normal Karyotype

23 homologous pairs



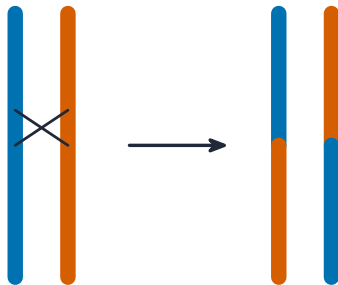
Meiosis

Two divisions of a diploid cell give four haploid, genetically different gametes. Meiosis I separates homologous pairs (halves the number). Meiosis II separates sister chromatids.

Crossing over in prophase I and independent assortment in metaphase I are the two sources of variation.

Variation from meiosis

crossing over



paired (a chiasma)
segments swapped
(new combinations)

Meiosis makes gametes
genetically different:

- **crossing over** – pairs swap segments
- **independent assortment**
– pairs line up randomly

Then **fertilisation** fuses any two
gametes at random.



The language of genetics

allele – one version of a gene

genotype vs **phenotype** – alleles vs features

dominant vs **recessive** – 1 copy shows vs needs 2

homozygous vs **heterozygous** – same vs different

A **test cross** with the recessive homozygote reveals an unknown genotype.

Four words the mark scheme insists on

codominant

both alleles show their effect **together** – not a blend, both present

linkage

genes on the same **autosome** tend to be inherited together, so the expected ratio breaks

epistasis

one gene affects whether another gene shows at all

When an observed ratio does not match the expected one, the **chi-squared** test decides whether the difference is chance or real – and linkage and epistasis are the two usual reasons it is real.

Crosses, and the genes behind two diseases

Monohybrid cross

A **monohybrid cross** follows **one** gene – one pair of alleles, four boxes in the grid.

Dihybrid cross

A **dihybrid cross** follows **two** genes at once: sixteen boxes, and the 9:3:3:1 ratio when both are heterozygous.

HBB

The **haemoglobin** gene: one base substitution gives **sickle cell anaemia**.

HTT

The huntingtin gene: a repeated triplet expands, and the result is **Huntington's disease** – dominant, and late-onset.

A named example is worth a mark. Pair each gene with its protein *and* the condition, as the syllabus table does.

Not every gene is switched on

- **structural genes** code for useful proteins – enzymes, haemoglobin
- **regulatory genes** decide whether other genes are switched on
- in eukaryotes, **transcription factors** bind to DNA and raise or lower **gene expression**

Gibberellin works exactly this way: it destroys the **DELLA** repressor proteins that were blocking a transcription factor, so the factor is freed and the gene turns on. The hormone does not switch the gene on – it removes what was holding it off.

Genetic crosses

parents: $Tt \times Tt$

	T	t
T	TT	Tt
t	Tt	tt

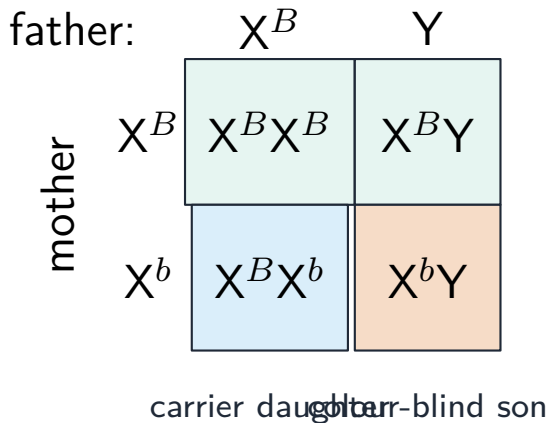
green = tall (has a T)

orange = short (tt)

ratio 3 tall : 1 short

A **Punnett square** shows how gametes combine – here $Tt \times Tt$ gives 3 tall : 1 short. A **monohybrid** follows one gene; a **dihybrid** two. The **chi-squared test** checks if results fit by chance.

Sex linkage



In **sex linkage** the gene is on the X chromosome, so the result differs by sex.

A carrier mother's **sons** may be colour-blind, while her daughters are only carriers.



From genes to phenotype

faulty **tyrosinase** → **albinism**

faulty factor VIII → **haemophilia**

faulty **haemoglobin** → **sickle cell**

faulty huntingtin → **Huntington's**

A gene codes for a protein, and the protein shapes the **phenotype** – so a faulty allele gives a faulty protein.

Gibberellin and stem height

In pea plants, the dominant allele *Le* codes for a working enzyme that makes **gibberellin** , so the plant grows tall by stem **elongation** .

The recessive allele *le* codes for a broken enzyme, so little gibberellin is made and the plant is short.

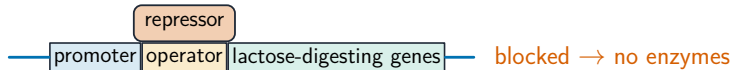
Gene control: the lac operon

The **lac operon** controls lactose digestion:

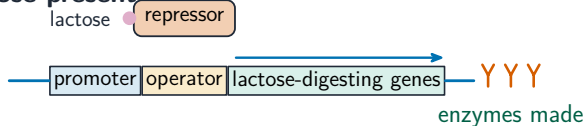
- no lactose → a **repressor** blocks the genes
- lactose pulls the repressor off → enzymes made (**inducible**)

Eukaryotes use **transcription factors** .

no lactose



lactose present



Switching a gene on when it is needed

The operon

In a **prokaryote** such as a bacterium, a group of genes – the **lac operon** – controls the digestion of lactose.

Off by default

A repressor protein binds the operator, so RNA polymerase cannot transcribe the genes. No lactose, no enzymes, no waste.

Lactose switches it on

Lactose binds the repressor and changes its shape, so it lets go, and transcription runs.

Two kinds of enzyme

An **inducible enzyme** is made only when it is needed; a **repressible enzyme** is normally made and can be switched off.

The cell is not deciding anything. A molecule changes a protein's shape, and the shape change is the decision.

The chi-squared test, and how gibberellin switches genes on

the chi-squared test

compares the numbers you actually counted (**observed**, O) with the numbers a hypothesis predicts (**expected**, E)

the statistic

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

the degrees of freedom

one fewer than the number of categories

the conclusion

if χ^2 exceeds the critical value at $p = 0.05$, the difference is **unlikely to be chance**

gibberellin

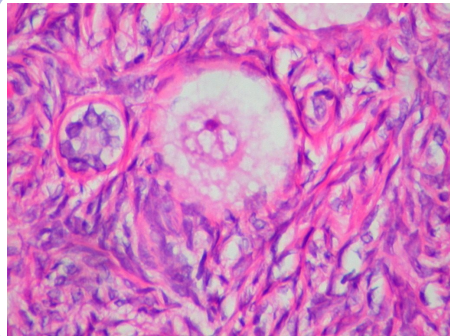
causes the breakdown of **DELLA** protein repressors

the effect

with the repressors gone, the genes they were blocking are transcribed – so the hormone acts by **removing a brake**

An **inducible enzyme** is made only when needed. Both gibberellin and the lac operon work by **lifting repression**, not by adding a signal to switch on.

Meiosis and how it creates variation



A human egg cell (ovum) – a haploid gamete produced by meiosis

The eight stages, and the genetics vocabulary

two divisions

meiosis has two, one after the other – so **eight named stages**

the names

prophase I, metaphase I, anaphase I, telophase I, then the same four numbered II

what division I does

separates the **homologous chromosomes** – this is where crossing over and independent assortment happen

what division II does

separates the sister chromatids, like mitosis

a locus

the position of a gene on a chromosome

monohybrid and dihybrid

crosses following one gene, and two

Division I is the reductive one, and it is where variation is generated. Division II adds no new combinations.

What the cell does at the start of meiosis I

Nuclear envelope

As in mitosis, the **nuclear envelope** breaks down and a **spindle** forms from the centrioles.

Pairing

Unlike mitosis, homologous chromosomes pair up – which is what makes crossing over possible.

Crossing over

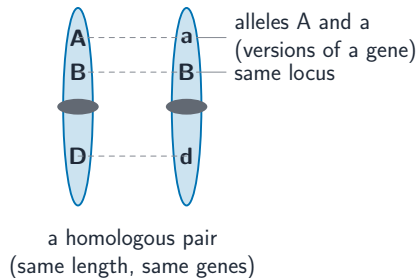
Chromatids exchange lengths at the chiasmata, so each chromatid ends up a new mixture of alleles.

Independent assortment

Each pair lines up at the equator independently of every other pair, giving 2^{23} combinations in a human gamete.

Two shuffles, then a third at fertilisation. Between them they explain almost all the variation in a sexually reproducing species.

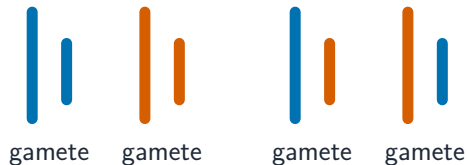
Meiosis and how it creates variation, continued



A homologous pair carries the same genes at the same loci, but the alleles (versions) may differ

Meiosis and how it creates variation, continued (2)

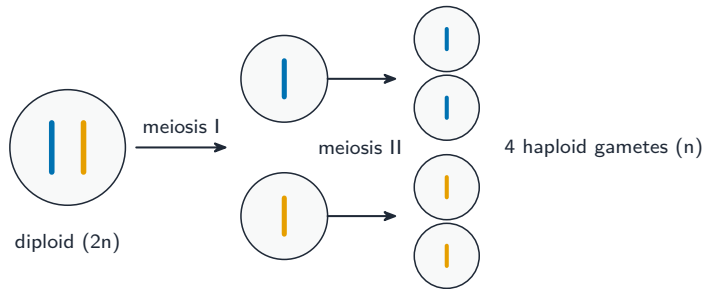
independent assortment



the pairs line up in a random order, so each gamete gets a random mix

Independent assortment: chromosome pairs line up in a random order, so gametes get many different mixes

Meiosis and how it creates variation, continued (3)



Two divisions halve the chromosome number: meiosis I separates the homologous pair, meiosis II separates the chromatids – giving four haploid gametes

Exam tips

- Set out a cross fully: **parental genotypes** → **gametes (in circles)** → **Punnett square** → **offspring ratio and phenotypes**.
- Use the **chi-squared** test to compare observed with expected; degrees of freedom = classes - 1, compare with 3.84 at $p = 0.05$.
- Recognise codominance, multiple alleles, sex linkage, epistasis and linkage – each alters the expected ratio.
- **Meiosis** creates variation by crossing over and independent assortment – state both.



Worked example – the chi-squared test

A $Tt \times Tt$ cross gives 160 offspring: 114 tall, 46 short. Does this fit 3 : 1?

- Expected: tall = $\frac{3}{4} \times 160 = 120$; short = $\frac{1}{4} \times 160 = 40$.
- $\chi^2 = \sum \frac{(O - E)^2}{E} = \frac{(114 - 120)^2}{120} + \frac{(46 - 40)^2}{40}$
- $= 0.30 + 0.90 = 1.20$
- Degrees of freedom = classes - 1 = 1; critical value at $p = 0.05$ is 3.84.
- $1.20 < 3.84 \Rightarrow$ difference is not significant; the data fit 3 : 1.

χ^2 below the critical value
 $\Rightarrow$ accept the null hypothesis (any difference is chance). Use raw counts, never percentages.

Topic 16 – key takeaways

1

Meiosis

halves chromosomes; makes variation

2

Terms

allele, dominant/recessive, genotype/phenotype

3

Crosses

Punnett squares; sex linkage differs by sex

4

Gene control

lac operon repressor; transcription factors

Check yourself

- 1 How many haploid gametes does meiosis make?
- 2 What does “heterozygous” mean?
- 3 $Tt \times Tt$ gives what phenotype ratio?
- 4 What pulls the repressor off the lac operon?

TT	Tt
Tt	tt

Answers 1. four 2. the two alleles are different 3. 3 tall : 1 short 4. lactose