Inheritance

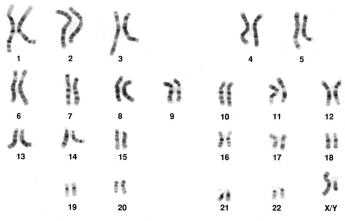
A-Level Biology ▪ Topic 16

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

1

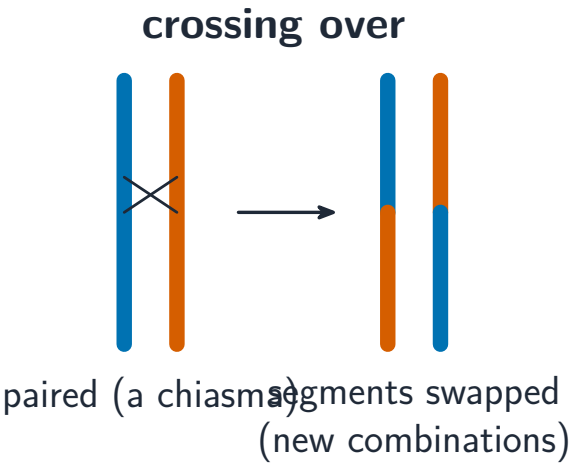
Meiosis

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



Meiosis makes gametes genetically different:

- **crossing over** 交叉互换 – pairs swap segments
- **independent assortment** 自由组合 – pairs line up randomly

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

2 Genetics

The language of genetics

allele 等位基因 – one version of a gene

genotype 基因型 vs **phenotype 表现型** – alleles vs features

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

homozygous 纯合 vs **heterozygous 杂合** – same vs different

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

Four words the mark scheme insists on

codominant 共显性 both alleles show their effect together – not a blend, both present

linkage 连锁 genes on the same **autosome** 常染色体 tend to be inherited together, so the expected ratio breaks

epistasis 上位性 one gene affects whether another gene shows at all

When an observed ratio does not match the expected one, the **chi-squared** 卡方检验 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** 单基因杂交 follows one gene – one pair of alleles, four boxes in the grid.
Dihybrid cross	A **dihybrid cross** 双基因杂交 follows two genes at once: sixteen boxes, and the $9:3:3:1$ ratio when both are heterozygous.
HBB	The **haemoglobin** 血红蛋白 gene: one base substitution gives **sickle cell anaemia** 镰状细胞贫血.
HTT	The huntingtin gene: a repeated triplet expands, and the result is **Huntington's disease** 亨廷顿病 – 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.

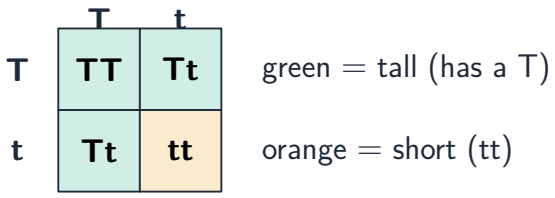
Not every gene is switched on

- **structural genes** 结构基因 code for useful proteins – enzymes, haemoglobin
- **regulatory genes** 调节基因 decide whether other genes are switched on
- in eukaryotes, **transcription factors** 转录因子 bind to DNA and raise or lower **gene expression** 基因表达

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 × Tt



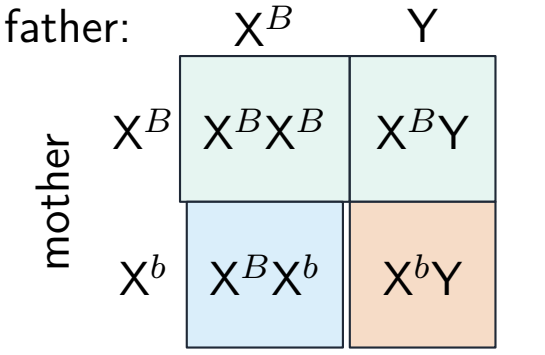
green = tall (has a T)

orange = short (tt)

ratio 3 tall : 1 short

A **Punnett square** 庞纳特方格 shows how gametes combine – here Tt × Tt gives 3 tall : 1 short. A **monohybrid** 单基因杂交 follows one gene; a **dihybrid** 双基因杂交 two. The **chi-squared test** 卡方检验 checks if results fit by chance.

Sex linkage



In **sex linkage** 伴性遗传 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.

3 Genes and control

From genes to phenotype

faulty **tyrosinase** 酪氨酸酶 → **albinism** 白化病

faulty **haemoglobin** 血红蛋白 → **sickle cell** 镰状细胞贫血

faulty factor VIII → **haemophilia** 血友病

faulty huntingtin → **Huntington's** 亨廷顿病

A gene codes for a protein, and the protein shapes the **phenotype** 表现型 – 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** 赤霉素, so the plant grows tall by stem **elongation** 伸长.

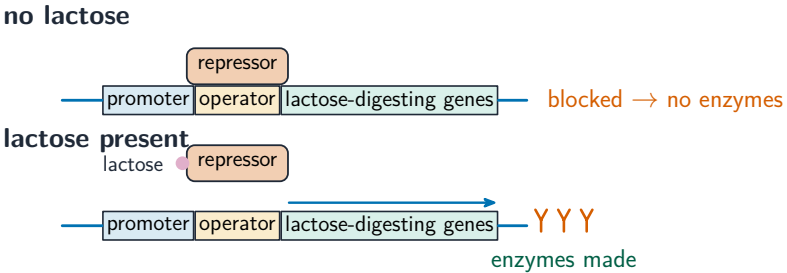
> The recessive allele *le* codes for a broken enzyme, so little gibberellin is made and the plant is short.

Gene control: the lac operon

The **lac operon** 乳糖操纵子 controls lactose digestion:

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

Eukaryotes use **transcription factors** 转录因子.



Switching a gene on when it is needed

The operon	In a **prokaryote** 原核生物 such as a bacterium, a group of genes – the **lac operon** 乳糖操纵子 – 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** 诱导酶 is made only when it is needed; a **repressible enzyme** 阻遏酶 is normally made and can be switched off.

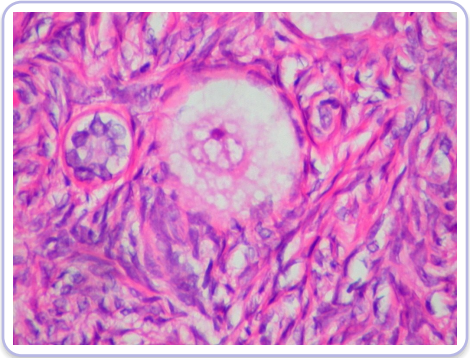
The cell is not deciding anything. A molecule changes a protein's shape, and the shape change is the decision.

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

the chi-squared test 卡方检验	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** 诱导酶 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** 同源染色体 – this is where crossing over and independent assortment happen
what division II does	separates the sister chromatids, like mitosis
a locus 基因座	the position of a gene on a chromosome
monohybrid 单因子杂交 and dihybrid 双因子杂交	crosses following one gene, and two

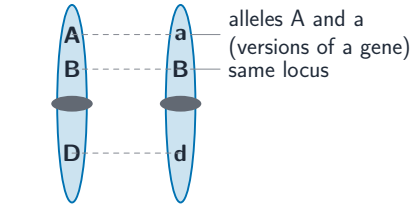
Division I is the reductive one, and it is where variation is generated. Division II adds no new combinations.

What the cell does at the start of meiosis I

Nuclear envelope	As in mitosis, the **nuclear envelope** 核膜 breaks down and a **spindle** 纺锤体 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
(same length, same genes)

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

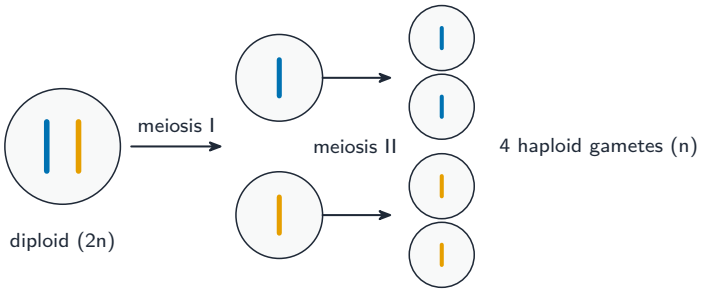
Meiosis and how it creates variation, continued (2)

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.

4 Recap

Worked example – the chi-squared test

A Tt × 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 × Tt gives what phenotype ratio?
4. What pulls the repressor off the lac operon?

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