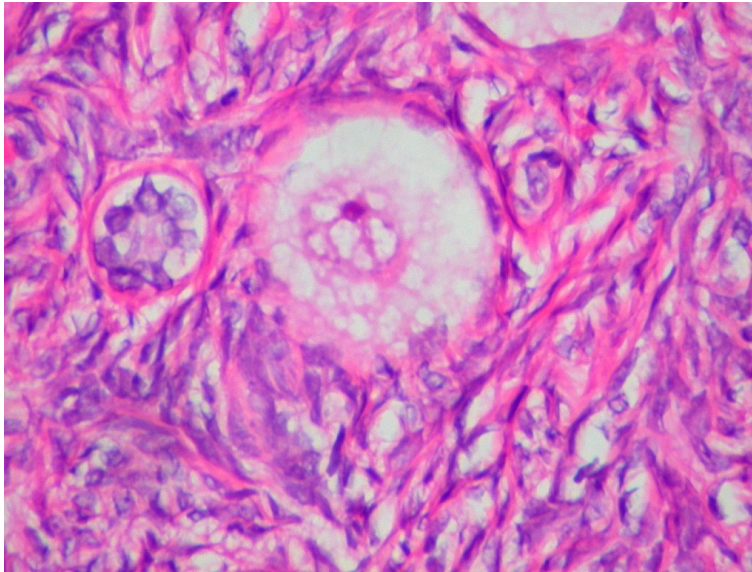


Inheritance

A-Level Biology

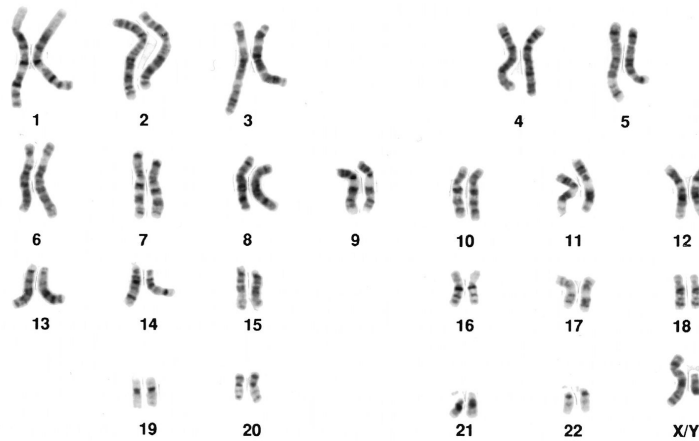
Meiosis and how it creates variation

A **diploid** 二倍体 cell ($2n$) has two full sets of **chromosomes** 染色体 — one set from each parent. A **haploid** 单倍体 cell (n) has just one set.



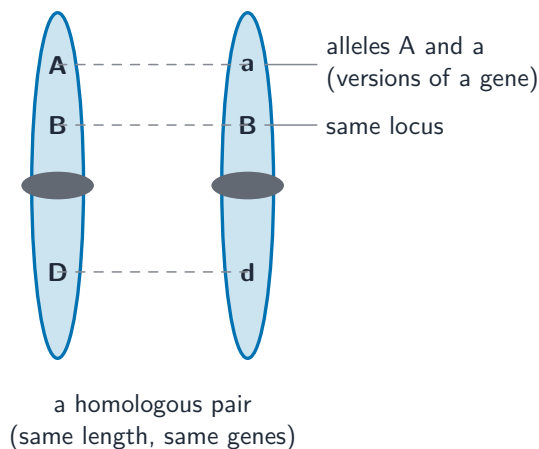
A human egg cell (ovum) — a haploid gamete produced by meiosis

Chromosomes come in **homologous chromosomes** 同源染色体 pairs: the two chromosomes in a pair are the same length and carry the same **genes** 基因 (though they may carry different versions of them).



Normal Karyotype

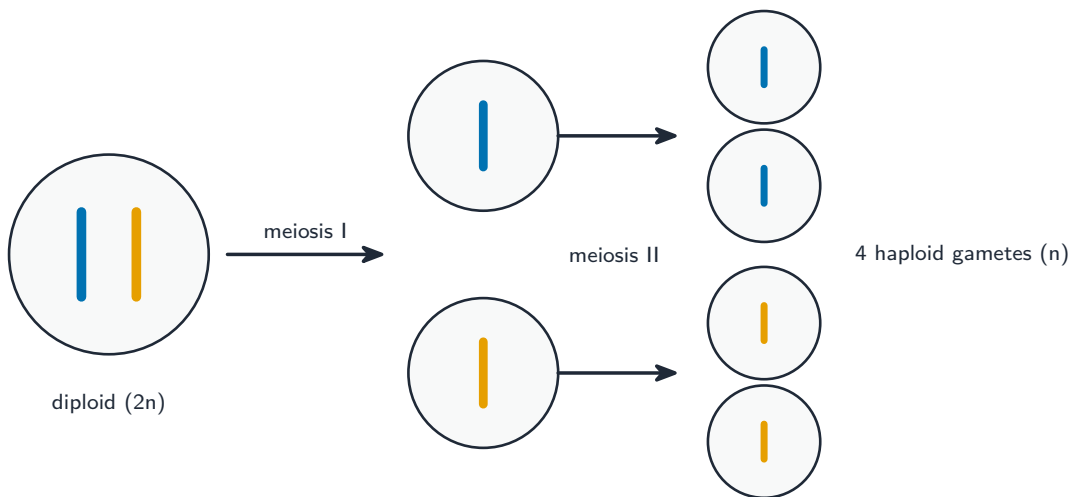
A human **karyotype**: the 46 chromosomes sorted into 23 homologous pairs (the last pair, X and Y, shows this is a male)



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

Gametes 配子 (sex cells) must be haploid. If they were diploid, the chromosome number would double at every generation. So gametes are made by a special division, **meiosis** 减数分裂, which halves the chromosome number.

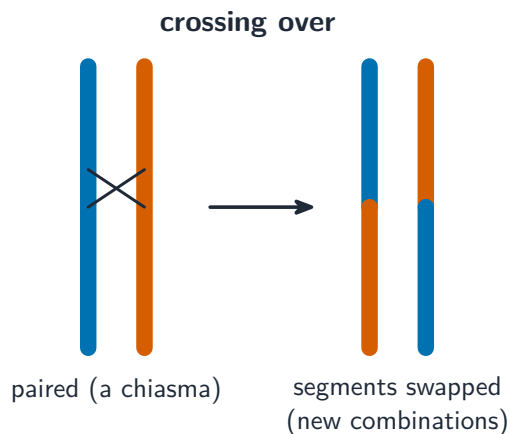
Meiosis has two divisions, one after the other, so there are eight named stages: prophase I, metaphase I, anaphase I and telophase I, then prophase II, metaphase II, anaphase II and telophase II. As in mitosis, the **nuclear envelope** 核膜 breaks down, a **spindle** 纺锤体 forms, and the chromosomes are moved by the spindle.



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

Meiosis makes the gametes **genetically different** from each other in two ways:

- **crossing over** 交叉互换 — homologous chromosomes swap matching pieces, mixing the alleles.



Crossing over: paired chromosomes swap matching segments at a chiasma, making new allele combinations

- **independent assortment** 自由组合 — the pairs line up in a random order, so each gamete gets a random mix of the parent's chromosomes.

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

Then at **fertilisation** 受精, any gamete can fuse with any other. This random fusion makes every new individual genetically different.

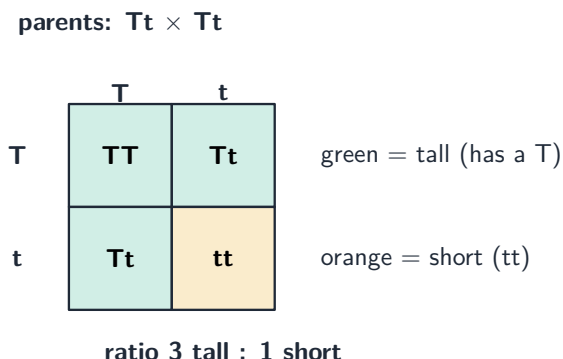
The language of genetics

You must know these terms exactly:

- a **gene** is a length of DNA that codes for a **protein** 蛋白质. Its position on a chromosome is its **locus** 基因座.
- an **allele** 等位基因 is one version of a gene.
- a **dominant** 显性 allele shows its effect even when only one copy is present; a **recessive** 隐性 allele only shows when two copies are present.
- **codominant** 共显性 alleles both show their effect together.
- the **genotype** 基因型 is the alleles an organism has; the **phenotype** 表现型 is the features you can see.
- **homozygous** 纯合 means the two alleles are the same; **heterozygous** 杂合 means they are different.
- a **test cross** 测交 crosses an organism with the recessive homozygote to find its unknown genotype.

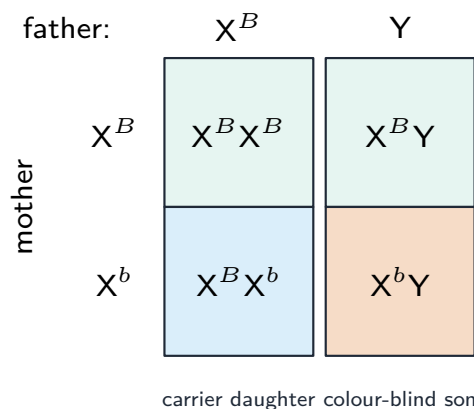
Genetic diagrams and crosses

You show a cross with a genetic diagram, often using a **Punnett square** 庞纳特方格 — a grid that shows all the ways the gametes can combine.



A Punnett square for $Tt \times Tt$: the offspring are 3 tall : 1 short (T is dominant)

- a **monohybrid cross** 单基因杂交 follows one gene; a **dihybrid cross** 双基因杂交 follows two genes at once.
- some genes have **multiple alleles** 复等位基因 (more than two versions in the population), such as the alleles for human blood groups.
- in **sex linkage** 伴性遗传, the gene is on the X chromosome, so the result is different for males and females.
- in **linkage** 连锁, genes on the same **autosome** 常染色体 (a non-sex chromosome) tend to be inherited together.
- in **epistasis** 上位性, one gene affects how another gene is shown.



Because the gene is on the X chromosome, a carrier mother's sons may be colour-blind while her daughters are only carriers — a different result by sex

The chi-squared test

The **chi-squared test** 卡方检验 compares the results you actually counted (the *observed* numbers, O) with the results you expected from the genetic diagram (the *expected* numbers, E). It tells you whether the difference is small enough to be due to chance, or large enough to mean something else is going on.

You add up, for every group, the squared difference between observed and expected, divided by the expected:

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

Then you compare this χ^2 value with a **critical value** from a table. The row you use is set by the *degrees of freedom* (the number of groups minus 1). If χ^2 is **less than** the critical value, the difference is not significant — the results fit the expected ratio, and any difference is just chance. If χ^2 is **greater than** the critical value, the difference is significant, so some other factor is involved.

Worked example. A Tt $\times$ Tt cross gives 160 offspring: 114 tall and 46 short. Test whether this fits the expected 3 : 1 ratio.

The expected numbers are $\frac{3}{4} \times 160 = 120$ tall and $\frac{1}{4} \times 160 = 40$ short. Then:

$$\chi^2 = \frac{(114 - 120)^2}{120} + \frac{(46 - 40)^2}{40} = \frac{36}{120} + \frac{36}{40} = 0.30 + 0.90 = 1.20.$$

There are 2 groups, so degrees of freedom = $2 - 1 = 1$. The critical value at $p = 0.05$ is 3.84. Because $1.20 < 3.84$, the difference is **not significant**: the results fit a 3 : 1 ratio, and the small difference is due to chance.

From genes to proteins to phenotype

A gene codes for a protein, and that protein affects the phenotype. If the gene is faulty, the protein is faulty, and the phenotype changes:

Gene	Protein	Effect of a faulty allele
TYR	the enzyme 酶 tyrosinase	albinism 白化病 (no pigment made)
HBB	haemoglobin 血红蛋白	sickle cell anaemia 镰状细胞贫血
F8	factor VIII (helps blood clot)	haemophilia 血友病
HTT	huntingtin	Huntington's disease 亨廷顿病

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

Not all genes are switched on all the time. Cells control which proteins they make.

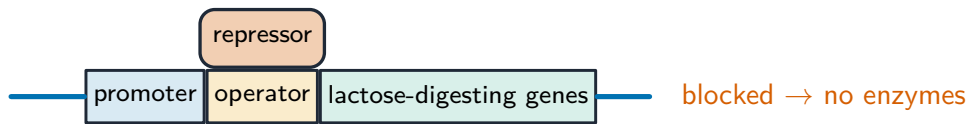
- **structural genes** 结构基因 code for useful proteins such as enzymes; **regulatory genes** 调节基因 control whether other genes are switched on.
- an **inducible enzyme** 可诱导酶 is made only when it is needed; a **repressible enzyme** 可抑制酶 is normally made but can be switched off.

The lac operon

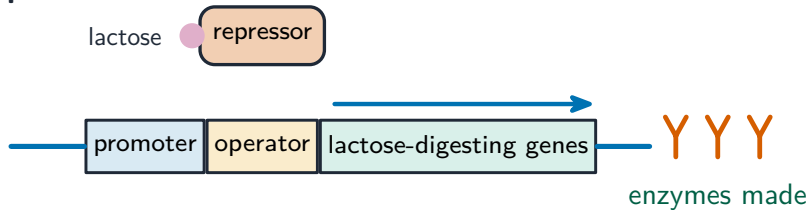
In a **prokaryote** 原核生物 such as a bacterium, a group of genes called the **lac operon** 乳糖操纵子 controls the digestion of lactose:

- when there is **no** lactose, a **repressor** 阻遏物 protein binds to the **operator** (a short control sequence within the operon) and blocks **transcription** 转录, so the lactose-digesting enzymes are not made.
- when lactose **is** present, it binds to the repressor and pulls it off. Transcription can now happen, and the enzymes are made. The enzymes are therefore inducible.

no lactose



lactose present



The lac operon: a repressor blocks the genes until lactose pulls it off, so the enzymes are inducible

Control in eukaryotes

In eukaryotes, **transcription factors** 转录因子 are proteins that bind to DNA and control **gene expression** 基因表达, by increasing or decreasing the rate of transcription.

Gibberellin switches genes on in this way: it causes the breakdown of **DELLA** protein repressors. These DELLA proteins normally block the factors that turn on transcription, so removing them lets those genes be expressed.

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.