

Edexcel GCSE Biology — Biology

Topic 3

Genetics

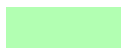
Specification 1BI0 · Pearson Edexcel

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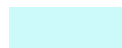
Revision notes for one topic, free to download.

These are the summary notes only. The full edition of this booklet also carries the worked examples, the required practicals and the formula sheets, with every topic in one place.

Colour Codes



Definitions



Diagrams



Notes and warnings



Sub-topic headings

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Define Sexual reproduction

- reproduction by the fusion of two gametes; offspring are genetically different.

Define Haploid

- one set of chromosomes (23 in humans); **diploid** — two sets (46).

Define Zygote

- the diploid cell formed when two gametes fuse.

Define Gene

- a section of a DNA molecule that codes for a specific protein.

Define Allele

- a different version of the same gene.

Define Homozygous

- two identical alleles of a gene; **heterozygous** — two different alleles.

Define Carrier

- a heterozygote who does not show a recessive condition but can pass the allele on.

Define Asexual reproduction

- reproduction from one parent by mitosis; offspring are genetically identical.

Define Meiosis

- cell division producing four genetically different haploid gametes from one diploid cell.

Define Gamete

- a sex cell, carrying one allele of each gene.

Define Chromosome

- a long DNA molecule in the nucleus, carrying many genes.

Define Genome

- the entire DNA of an organism.

Define Dominant

- an allele expressed even if only one copy is present; **recessive** — expressed only when two copies are present.

Define Genotype

- the alleles an organism has; **phenotype** — the characteristics it shows.

Define Mutation

- a change in the sequence of bases in DNA.

Asexual and sexual reproduction (3.1B–3.2B) BIOLOGY ONLY

BIOLOGY ONLY Asexual reproduction uses one parent and no gametes: cells divide by **mitosis**, giving genetically identical **clones**. In **sexual reproduction** two gametes fuse, mixing alleles from both parents.

	Asexual	BIOLOGY ONLY BIOLOGY ONLY Sexual
Parents	One	Two
Cell division	Mitosis only	Meiosis to make gametes, then mitosis
Offspring	Genetically identical clones	Genetically different
Advantages	No need to find a mate; rapid reproductive cycle	Variation , so if the environment changes some individuals are likely to survive
Disadvantages	No variation , so one disease could wipe out the population	Time and energy spent finding a mate ; slower reproductive cycle

Meiosis and gametes (3.3)

- One diploid cell divides to produce **four daughter cells**.
- Each has **half the number of chromosomes** of the parent — they are **haploid**.
- The four gametes are **genetically different** from each other and from the parent cell.
- At **fertilisation** two gametes fuse, restoring the **diploid** number ($23 + 23 = 46$). The zygote divides by mitosis to form an embryo.

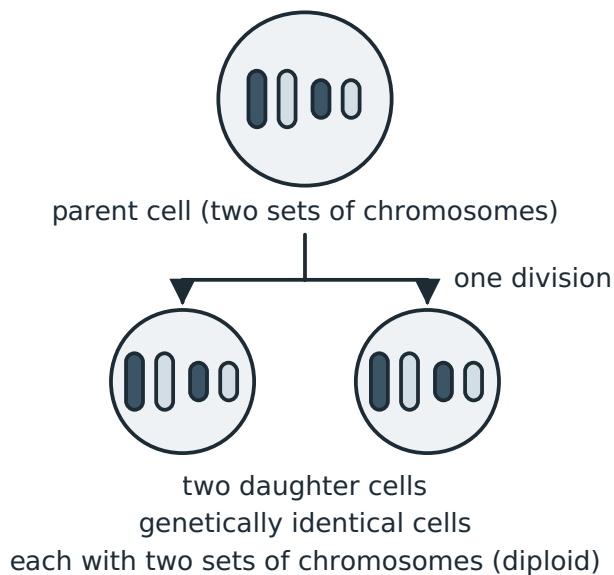
NOTE

The **stages of meiosis** are **not required** by this specification — only the four points above.

	Mitosis	Meiosis
Daughter cells	2	4
Chromosome number	Same as parent (diploid)	Half that of the parent (haploid)
Genetically	Identical to parent and each other	Different from parent and each other
Where	All body cells	Reproductive organs only
Purpose	Growth, repair, asexual reproduction	Making gametes for sexual reproduction

Mitosis and meiosis compared

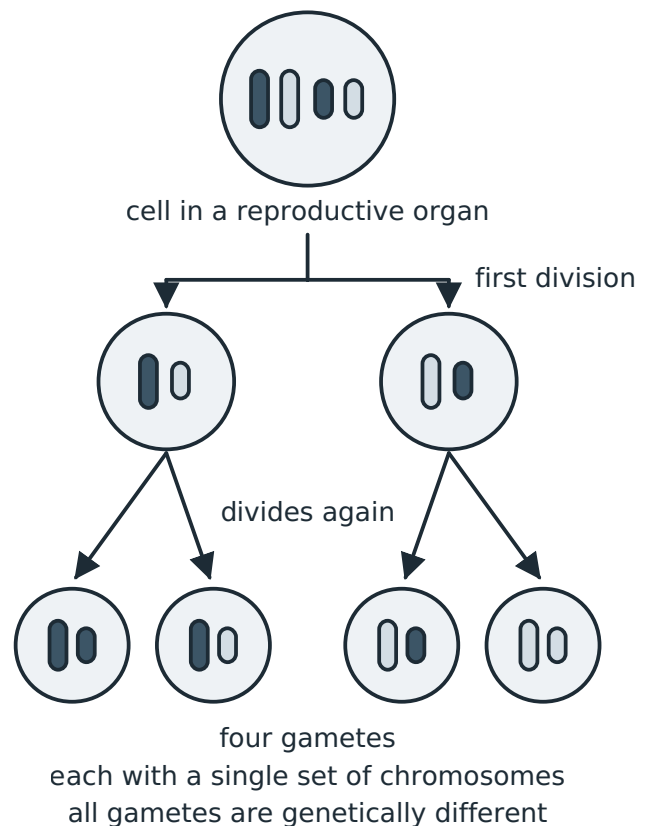
MITOSIS



 dark = chromosome from one parent
 light = chromosome from the other parent

Asexual reproduction involves only mitosis, so the offspring are genetically identical (clones).

MEIOSIS



Knowledge of the stages of meiosis is not required.

DNA, genes and the genome (3.4–3.6)

DNA is a polymer (3.4)

- **a** Two strands coiled to form a **double helix**.
- **b** The strands are linked by **complementary base pairs** joined by **weak hydrogen bonds**: **A pairs with T, C with G**. Weak bonds let the strands separate when DNA is copied.
- **c** DNA is made of **nucleotides** — a **sugar** and a **phosphate group** with **one of four bases** attached to the sugar. Sugars and phosphates form each strand's backbone.

Genes and the genome (3.5). A **gene** is a section of a DNA molecule that **codes for a specific protein**; the **genome** is the **entire DNA of an organism**. Size order: nucleus → chromosome → DNA molecule → gene → base.

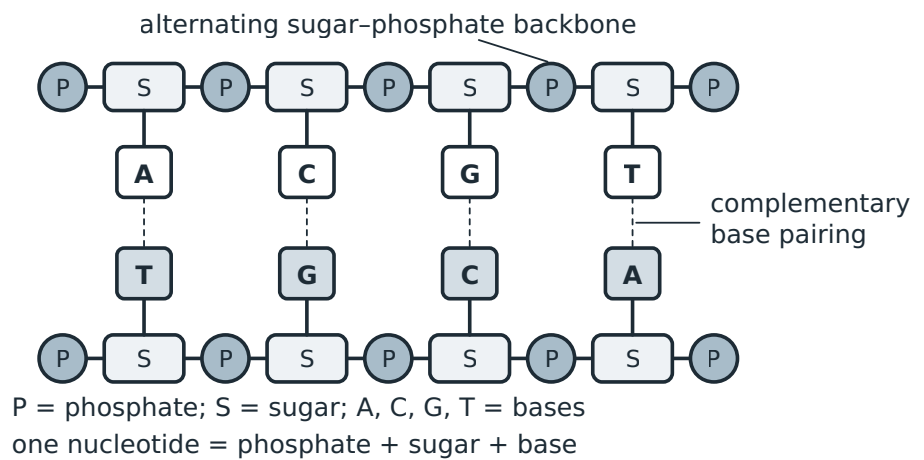
The structure of DNA



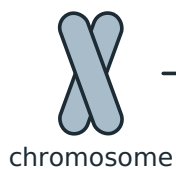
DNA double helix
two strands

opened out →

The two strands opened out



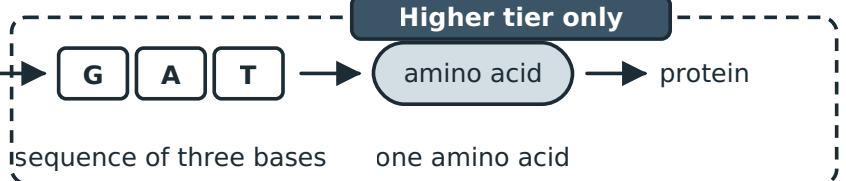
In the complementary strands a C is always linked to a G on the opposite strand, and a T to an A.



chromosome



gene



Each gene codes for a particular sequence of amino acids, to make a specific protein.

ANALOGY

The genome is a recipe book for building an organism: each chromosome is a volume, each gene a recipe for one protein, and A, T, C and G are the only four letters it is written in. The **order** of the letters carries the meaning — change one letter and you may change the dish, or nothing at all.

Extracting DNA from fruit (3.6)

1. **Mash** the fruit to break down the cell walls.
2. Add **detergent** (breaks down cell and nuclear membranes, releasing DNA) and **salt** (makes the DNA clump together).
3. **Filter** to remove insoluble cell debris.
4. Add ice-cold **ethanol** down the side of the tube. DNA is insoluble in it, so **precipitates** as white stringy strands at the boundary.

Protein synthesis (3.7B–3.10B) BIOLOGY ONLY

HT ONLY

BIOLOGY ONLY HT ONLY The genetic code (3.7B). The **order of bases** in a section of DNA decides the **order of amino acids** in the protein. The chain then **folds** into a specifically shaped protein, such as an enzyme whose active site lets it do its job.

BIOLOGY ONLY HT ONLY Stages of protein synthesis (3.8B)

Transcription (in the nucleus)

- **a RNA polymerase** binds to **non-coding DNA** in front of a gene.
- **b** It produces a **complementary mRNA strand** from the **coding DNA** of the gene; the mRNA leaves the nucleus.

Translation (at the ribosome)

- **c** The **mRNA attaches to a ribosome**.
- **d** The mRNA bases are read in **triplets called codons**, each coding for a specific amino acid.
- **e tRNA transfers amino acids to the ribosome**, each carrying the amino acid matching the codon being read.
- **f** The amino acids are **linked to form a polypeptide**, which folds into the finished protein.

BIOLOGY ONLY HT ONLY Variants in non-coding DNA (3.9B). A variant in the non-coding DNA in front of a gene changes how well **RNA polymerase binds**. Poorer binding means **less protein is produced**, better binding more. Changing the **quantity** of a protein can change the phenotype.

BIOLOGY ONLY HT ONLY Variants in coding DNA (3.10B). A variant in coding DNA alters the **sequence of amino acids**, so the protein folds into a different shape. In an enzyme this can change the active site so the substrate no longer fits, altering the **activity of the protein** and so the phenotype.

The work of Mendel (3.11B) BIOLOGY ONLY

BIOLOGY ONLY Gregor Mendel bred pea plants in the 1850s–60s, crossing pure-breeding plants that differed in one characteristic and counting the offspring over several generations.

- One form disappeared in the first generation and reappeared in about a quarter of the second — a **3 : 1 ratio**.
- He concluded that characteristics are controlled by separate "units" (now **genes**), inherited one from each parent, and that one unit can be **dominant** over the other.
- **Why it was not accepted:** chromosomes and DNA were unknown, so there was no mechanism to explain his units. His work was recognised only after his death, once chromosome behaviour in meiosis was seen to match his results.

Alleles and genetic terms (3.12–3.13)

Why inherited characteristics differ (3.12). Body cells carry two copies of each gene, one from each parent. The two copies may be different versions — **alleles**. Which alleles are inherited, and whether they are dominant or recessive, decides the characteristic shown.

Statement 3.13 requires you to explain **chromosome, gene, allele, dominant, recessive, homozygous, heterozygous, genotype, phenotype, gamete** and **zygote** — all defined at the top of this topic. Write dominant alleles as capitals, recessive as lower case.

ANALOGY

Dominant and recessive alleles are a shout and a whisper in one room. If anyone shouts (**dominant**) that is all you hear. You hear the whisper (**recessive**) only when nobody is shouting — when both alleles are recessive. The whisper is still there in a heterozygote and can be passed on; it is simply not heard.

Monohybrid inheritance, pedigrees and sex determination (3.14–3.16)

Monohybrid inheritance — inheritance of a characteristic controlled by a **single gene** with two alleles — is worked out with **genetic diagrams, Punnett squares** and **family pedigrees** (3.14). To draw a Punnett square, write both **parent genotypes**, then each parent's **gametes** along the top and side; combine row and column alleles in each box; convert the **genotypes** to **phenotypes**, then to a ratio or probability.

How to set out a Punnett square

Parents: Bb × Bb

both parents heterozygous, both with black fur

gametes from parent 1

		gametes from parent 1	
		B	b
gametes from parent 2	B	BB black	Bb black
	b	Bb black	bb brown

Genotype ratio BB : Bb : bb = 1 : 2 : 1

Phenotype ratio black : brown = 3 : 1

B = dominant allele (black fur): always expressed

b = recessive allele (brown fur): needs two copies to be expressed

Probability of brown fur = 1 in 4 (25%)

	T	t
t	Tt	tt
t	Tt	tt

	B	b
B	BB	Bb
b	Bb	bb

WATCH OUT

Predicted ratios are only what is **expected**. Fertilisation is random and each fertilisation is independent — parents with three affected children still have a ¼ chance for the next.

Inherited disorders and carriers. Many single-gene disorders are caused by a **recessive** allele. **Cystic fibrosis** is a disorder of cell membranes causing thick, sticky mucus in the lungs and digestive system. Only homozygous recessive individuals have it; a heterozygote is a **carrier**.

Inheritance of a recessive disorder such as cystic fibrosis

Cystic fibrosis

caused by a recessive allele

Parents: Ff × Ff — both carriers
gametes

gametes		F	f
	F	FF unaffected	Ff carrier
	f	Ff carrier	ff affected

1 in 4 (25%) has cystic fibrosis
2 in 4 (50%) are carriers
1 in 4 (25%) is unaffected

F = dominant allele (unaffected)
f = recessive allele (cystic fibrosis)

Cystic fibrosis is a disorder of cell membranes.

Polydactyly

caused by a dominant allele

Parents: Dd × dd — one affected parent
gametes

gametes		D	d
	d	Dd affected	dd unaffected
	d	Dd affected	dd unaffected

2 in 4 (50%) have polydactyly
2 in 4 (50%) are unaffected
so a 1 in 2 chance of being affected

D = dominant allele (polydactyly)
d = recessive allele (unaffected)

Polydactyly is having extra fingers or toes.

	F	f
F	FF	Ff
f	Ff	ff

Family pedigrees (3.14, 3.16)

- Squares are males, circles females; shaded symbols are affected; horizontal lines join parents, vertical lines lead to their children.
- The allele is **recessive** if two unaffected parents have an **affected child** — that child is homozygous recessive, so both parents must be carriers.
- The allele is **dominant** if every affected child has an affected parent and the condition appears in every generation. You can then deduce unknown genotypes and probabilities.

Sex determination (3.15). The 23rd pair of chromosomes are the **sex chromosomes**: **XX** in females, **XY** in males. All eggs carry **X**; sperm carry **X** or **Y**, so the **sperm** determines the sex of the offspring.

Parents: XX × XY

mother — female (XX) father — male (XY)

		gametes from father	
		X	Y
gametes from mother —	X	XX female	XY male
	X	XX female	XY male

Ratio female : male = 1 : 1

2 in 4 (50%) female, 2 in 4 (50%) male

a 50 : 50 chance of a male or a female offspring

Ordinary human body cells contain 23 pairs of chromosomes.
22 pairs control characteristics only; one pair determines sex.

	X (mother)	X (mother)
X (father)	XX	XX
Y (father)	XY	XY

Codominance and sex linkage (3.17B–3.18B) BIOLOGY ONLY

BIOLOGY ONLY ABO blood groups (3.17B) are controlled by a gene with **three alleles** — **multiple alleles** — although each person has only two. **IA** and **IB** are **codominant**, so if both are present **both are expressed**; **IO** is recessive to both.

Blood group	Genotypes
A	IAIA or IAIO
B	IBIB or IBIO
AB	IAIB (codominance)
O	IOIO

Codominance and the ABO blood group alleles

Group	Genotypes	Antigens
A	$I^A I^A$ or $I^A I^O$	A only
B	$I^B I^B$ or $I^B I^O$	B only
AB	$I^A I^B$	A and B
O	$I^O I^O$	none

I^A and I^B are both dominant to I^O , so $I^A I^O$ gives group A and $I^B I^O$ gives group B.

Group O makes no antigens; group AB makes both, because the two alleles are codominant.

Worked cross: group A × group B

Mother: $I^A I^O$ (group A)

Father: $I^B I^O$ (group B)

		father's gametes	
		I^B	I^O
mother's gametes	I^A	$I^A I^B$ group AB	$I^A I^O$ group A
	I^O	$I^B I^O$ group B	$I^O I^O$ group O

All four blood groups are possible — each has a 1 in 4 (25%) chance.

Codominance and multiple alleles

In group AB neither allele is dominant over the other. I^A and I^B are codominant, so both are expressed and the red blood cells carry both A antigens and B antigens.

The ABO gene has three alleles (I^A , I^B and I^O) — this is called multiple alleles — but any one person carries only two of them, one inherited from each parent.

HIGHER TIER

BIOLOGY ONLY HT ONLY Sex-linked genetic disorders (3.18B)

are caused by alleles on the **X chromosome**;

the smaller Y carries no matching allele.

- A female (**XX**) has two copies, so a recessive faulty allele on one X is masked by the normal allele on the other — she is a **carrier**.
- A male (**XY**) has only one copy, so a single recessive faulty allele **is** expressed — which is why haemophilia and red-green colour blindness are commoner in males.
- Write the allele as a superscript on the X: $XHx \times XHY$ gives $\frac{1}{4}$ carrier daughters, $\frac{1}{4}$ unaffected daughters, $\frac{1}{4}$ unaffected sons, $\frac{1}{4}$ affected sons — **half the sons affected, no daughters**.

Variation, mutation and the Human Genome Project (3.19–3.23)

Polygenic inheritance (3.19). Most phenotypic features result from **multiple genes**, not single-gene inheritance — for example height, skin colour and body mass.

Causes of variation (3.20)

- **a Genetic variation** — characteristics differing as a result of **mutation** and **sexual reproduction**, which shuffles alleles. It is inherited.
- **b Environmental variation** — characteristics caused by an organism's **environment (acquired characteristics)**, e.g. a scar, a suntan, a shaded plant growing tall and pale. Not inherited.
- Most characteristics are affected by **both**, e.g. height depends on genes and on diet.

Mutation and variation in populations (3.22–3.23)

- There is usually **extensive genetic variation within a population**, arising through **mutations** — changes in the sequence of bases in DNA.
- **Most mutations have no effect on the phenotype; some have a small effect; rarely, a single mutation significantly affects the phenotype**, e.g. producing a non-functional enzyme. Natural selection acts on this variation.

The Human Genome Project (3.21) determined the **order of all the bases in the human genome** and identified its genes.

Outcomes and applications in medicine:

- **Predicting and preventing disease** — genes raising the risk of conditions such as some cancers can be identified, so those at risk are screened or advised to change lifestyle.
- **Testing and diagnosis** — faster, more reliable testing for inherited disorders and carriers.
- **New treatments** — knowing which faulty protein causes a disorder lets drugs be designed against it, and opens up gene therapy.
- **Personalised medicine** — drugs and doses matched to an individual's genome.
- **Issues** — the information could be misused by employers or insurers, and knowing your risk of an incurable disease causes stress.

Use of mathematics in this topic

- Use **direct proportions and simple ratios**, and **probability**, in genetic crosses.
- Calculate **arithmetic means**, e.g. mean height in a study of continuous variation.
- Use **estimations**, and explain when they should be used.
- **Translate between numerical and graphical forms**; interpret graphs, charts and tables — histograms for continuous variation, bar charts for discontinuous variation.