

Genetics revision notes

Genetics explains how characteristics are passed from parents to offspring. This topic covers reproduction and meiosis, the structure of DNA, how genes code for proteins, patterns of inheritance, and the causes of variation.

For Edexcel GCSE Biology 1BI0 and Combined Science 1SC0. **Triple only** is not in Combined Science. **Higher tier** is only on the Higher paper.

1 Reproduction and meiosis Spec 3.1B–3.3

Asexual reproduction Spec 3.1B Triple only

In **asexual reproduction** there is only one parent. The offspring are genetically identical to the parent, so there is **no variation** in the population.

Asexual reproduction: advantages and disadvantages

	Point	Why it matters
Advantage	No need to find a mate	Saves time and energy. Useful when individuals are far apart.
Advantage	Rapid reproductive cycle	Many offspring are made quickly, so the population can grow fast.
Disadvantage	No variation in the population	All the individuals have the same weaknesses. A new disease or a change in the environment could kill them all.

The key idea is the trade-off. Asexual reproduction is quick and needs no partner, but the lack of variation makes the population vulnerable if conditions change.

Exam tip: Do not just write that there is no variation. Link it to a consequence: for example, none of the population would be resistant to a new disease.

Sexual reproduction Spec 3.2B Triple only

In **sexual reproduction** two parents each produce gametes. The gametes fuse at fertilisation, so the offspring inherit a mixture of alleles from both parents. This produces **variation** in the population.

Sexual reproduction: advantages and disadvantages

	Point	Why it matters
Advantage	Variation in the population	If the environment changes or a disease appears, some individuals are likely to have characteristics that let them survive and reproduce.
Disadvantage	Requirement to find a mate	This takes time and energy, and may not always be possible.

Common mistake: Sexual reproduction does not make identical offspring, and the offspring are not identical to either parent. Always say they are genetically different.

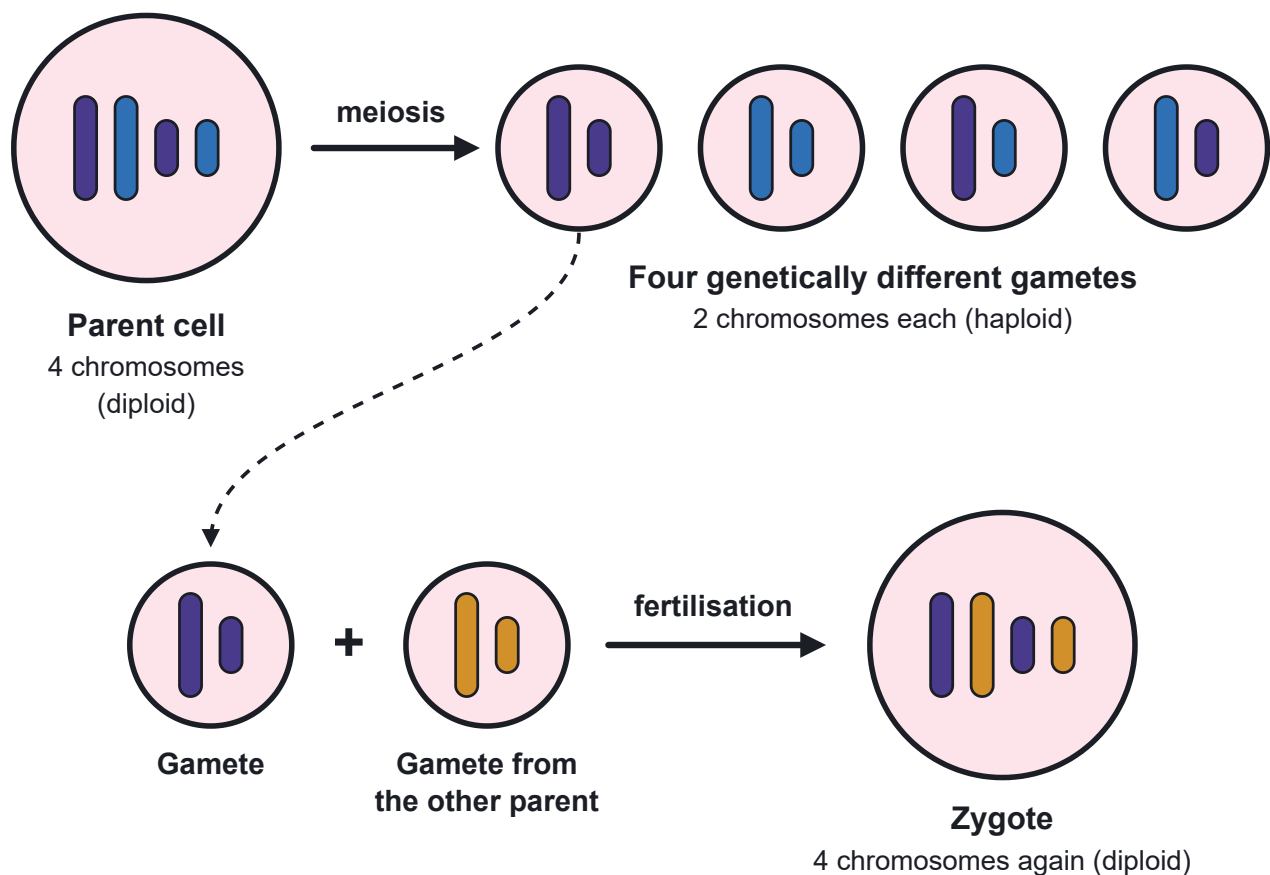
Meiosis Spec 3.3

Meiotic cell division (meiosis) is the type of cell division that makes gametes. In humans it happens in the reproductive organs.

- One parent cell divides to produce **four daughter cells**.
- Each daughter cell has **half the number of chromosomes** of the parent cell.
- The daughter cells are **genetically different** from each other and from the parent cell.
- The daughter cells are **haploid gametes**, with one set of chromosomes.

A human body cell has 46 chromosomes. Meiosis makes sperm and egg cells with 23 chromosomes each. At fertilisation the two gametes fuse and the zygote has 46 chromosomes again, with half from each parent.

Body cells and gametes		
	Body cell (diploid)	Gamete (haploid)
Chromosomes in a human cell	46	23
Genetically identical to the others?	Yes, if made by mitosis	No, each gamete is different
Made by	Mitosis	Meiosis



Meiosis halves the chromosome number; **fertilisation** restores it in the zygote.

Exam tip: The stages of meiosis are not required. Learn the outcome: four genetically different haploid gametes, each with half the number of chromosomes.

Quick check

- 1 . Give one advantage and one disadvantage of asexual reproduction.
- 2 . Give one advantage and one disadvantage of sexual reproduction.
- 3 . How many daughter cells does one meiotic division produce?
- 4 . How many chromosomes are in a human gamete?

2 DNA and the genome Spec 3.4-3.6

The structure of DNA Spec 3.4

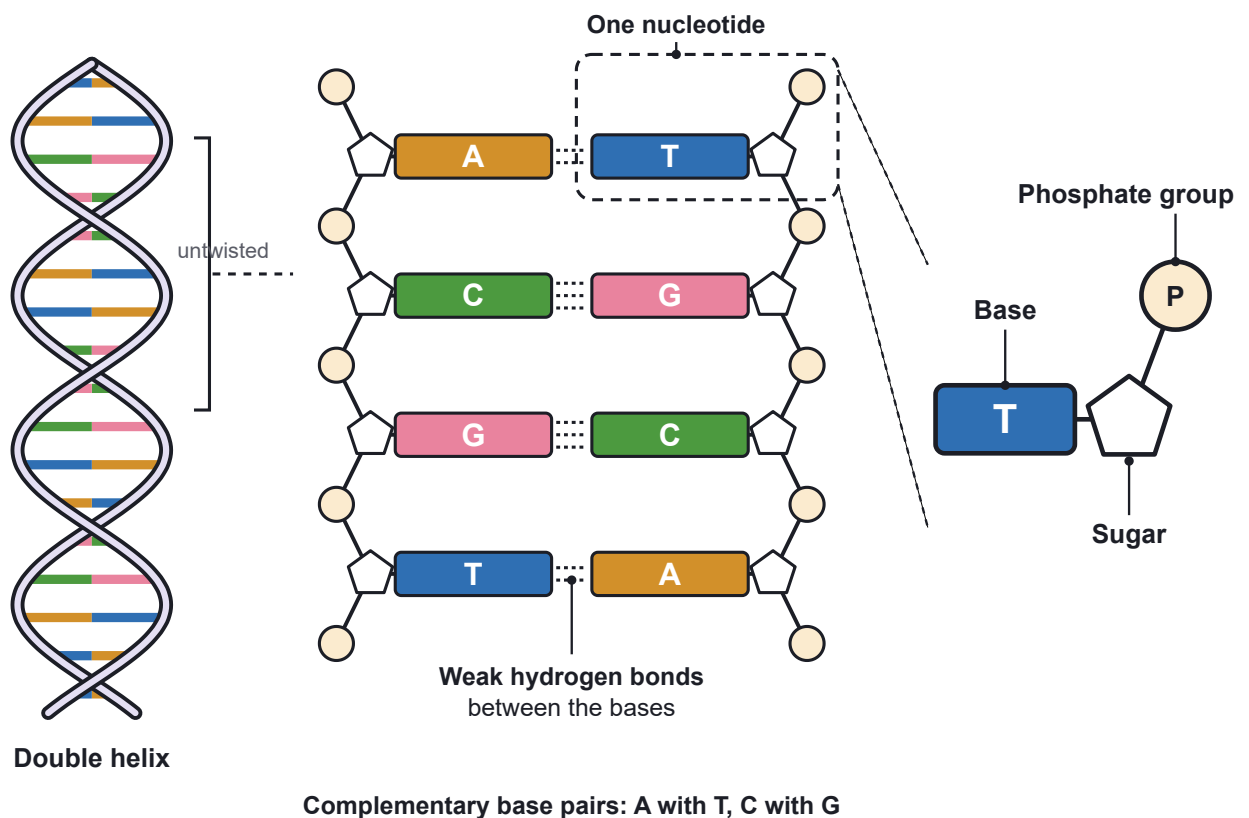
DNA is a **polymer** made up of many repeating units called **nucleotides**. A polymer is a long molecule made of many similar smaller units joined together.

- DNA is made of two strands coiled to form a **double helix**.
- Each nucleotide consists of a sugar and a phosphate group, with one of four different bases attached to the sugar.
- The two strands are linked by a series of **complementary base pairs**, joined together by **weak hydrogen bonds**.

The four bases and how they pair

Base	Pairs with
Adenine (A)	Thymine (T)
Thymine (T)	Adenine (A)
Cytosine (C)	Guanine (G)
Guanine (G)	Cytosine (C)

Because A always pairs with T and C always pairs with G, the base sequence on one strand tells you the sequence on the other. If one strand reads A T G C, the complementary strand reads T A C G.



DNA is a double helix of nucleotides; the bases pair **A with T** and **C with G**, held by weak hydrogen bonds.

Common mistake: The bonds between the two strands are weak hydrogen bonds. They are between the bases, not between the sugar and phosphate groups.

The genome and genes Spec 3.5

Genome	The entire DNA of an organism.
Gene	A section of a DNA molecule that codes for a specific protein.

The genome includes all of the genes of an organism. A gene is a short section of the DNA. Each gene has a different sequence of bases and so codes for a different protein.

Exam tip: Use the exact wording. A gene codes for a specific protein, and the genome is the entire DNA, not just the genes.

Extracting DNA from fruit Spec 3.6

DNA can be extracted from the cells of fruit such as strawberries or kiwi fruit using simple equipment. Each step has a purpose.

1. Mash the fruit in a beaker. This breaks the fruit into small pieces and separates the cells.
2. Add a mixture of detergent and salt, and stir. The detergent breaks down the cell membranes (and the nuclear membranes) so the DNA is released. The salt helps the DNA to clump together.
3. Filter the mixture. This removes the solid pieces of cell, and the filtrate contains the DNA.
4. Carefully add ice-cold ethanol so that it forms a layer on top of the filtrate.
5. Look at the boundary between the layers. DNA is not soluble in cold ethanol, so it comes out of solution as white, stringy strands. These can be lifted out with a glass rod.

Exam tip: In a 'why' question, give the purpose of each reagent: detergent breaks down membranes, salt helps the DNA clump together, and ice-cold ethanol makes the DNA come out of solution (precipitate) so it can be seen.

Common mistake: Do not say that you can see the double helix. You see a mass of thousands of DNA strands clumped together.

Quick check

1. What are the two strands of DNA coiled into?
2. Which bases pair together in DNA?
3. What is a gene?
4. What is the purpose of the detergent when extracting DNA from fruit?

3 Protein synthesis and gene variants Spec 3.7B-3.10B Triple only Higher tier

From base order to protein shape Spec 3.7B

A gene is a section of DNA that codes for a specific protein. Proteins are made of chains of amino acids. The order of bases in a section of DNA decides the order of amino acids in the protein.

1. The sequence of bases in the gene decides the order of amino acids.
2. The chain of amino acids folds up.
3. The way the chain folds depends on the order of the amino acids, so it produces a specifically shaped protein.

The shape of a protein matters because it decides what the protein can do. For example, an enzyme has an active site with a shape that fits its substrate. A different order of bases would give a different order of amino acids, a differently folded protein and so a different shape.

Exam tip: Show the full chain of ideas in your answer: order of bases, then order of amino acids, then folding, then specific shape, then function.

Protein synthesis: transcription and translation Spec 3.8B

Protein synthesis has two main stages. In transcription, a copy of the gene is made as a strand of mRNA. In translation, the mRNA is used to build a polypeptide at a ribosome.

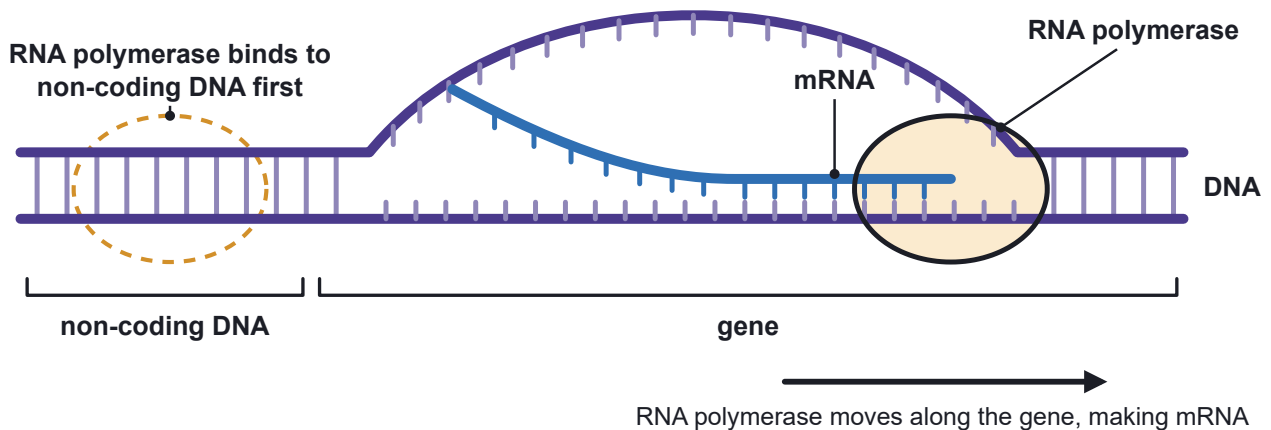
Transcription (in the nucleus)

1. RNA polymerase binds to the non-coding DNA located in front of a gene.
2. RNA polymerase produces a complementary mRNA strand from the coding DNA of the gene.
3. The mRNA moves out of the nucleus into the cytoplasm.

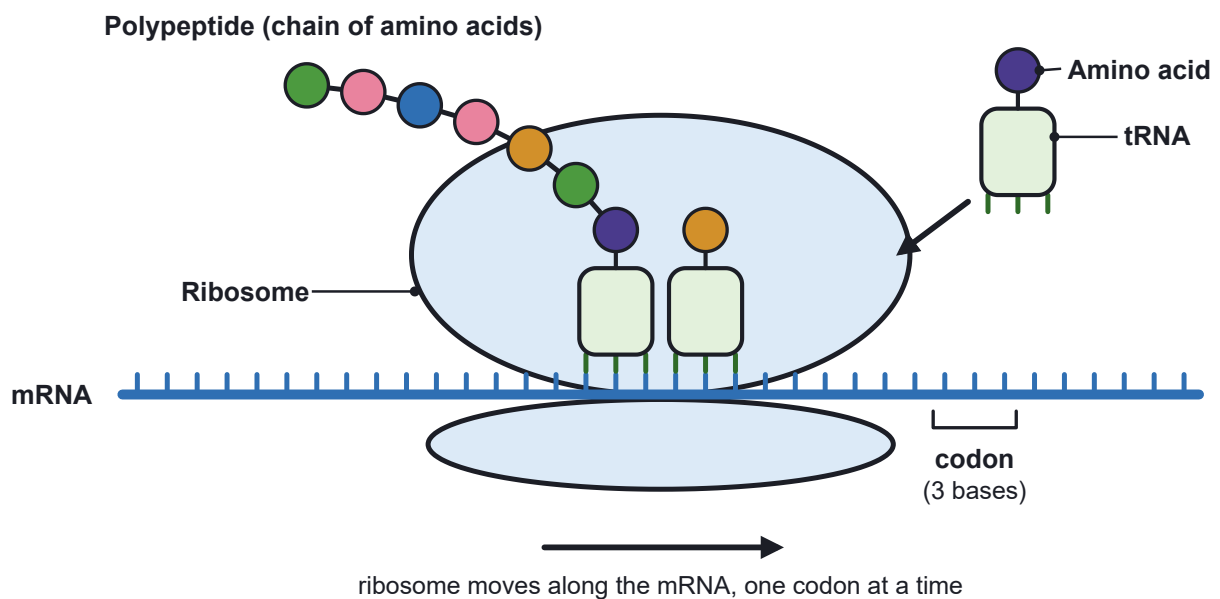
Translation (at a ribosome)

1. The mRNA attaches to a ribosome.
2. The mRNA is read in triplets of bases called codons. Each codon codes for a specific amino acid.
3. tRNA molecules transfer amino acids to the ribosome. Each tRNA carries the amino acid that matches a codon on the mRNA.
4. The amino acids are linked together to form a polypeptide, which folds into a protein.

1 Transcription (in the nucleus)



2 Translation (at a ribosome)



Transcription makes mRNA from DNA; **translation** makes a polypeptide from mRNA at a ribosome.

Common mistake: Do not mix up the two stages. Transcription makes mRNA from DNA. Translation makes a polypeptide from mRNA.

Genetic variants in non-coding DNA Spec 3.9B

A **genetic variant** is a difference in the DNA sequence. Variants in the **non-coding DNA** in front of a gene can affect the **phenotype** without changing the protein itself.

1. The variant is in the non-coding DNA, where RNA polymerase binds.
2. This can change how well RNA polymerase **binds**.
3. More or less mRNA is made, so the **quantity of protein** produced is altered.
4. The change in the amount of protein can change the phenotype.

Exam tip: A variant in non-coding DNA changes how much protein is made. It does not change the amino acid sequence of the protein.

Genetic variants in coding DNA Spec 3.10B

Variants in the **coding DNA** of a gene can change the base sequence that codes for the protein.

- 1 . A variant in the coding DNA can alter the **sequence of amino acids**.
- 2 . The polypeptide may then fold into a different shape.
- 3 . A protein with a different shape may have different **activity**. For example, an enzyme's active site may no longer fit its substrate.
- 4 . The change in the activity of the protein can affect the phenotype.

Where a variant is and what it changes

Where the variant is	What changes	Effect on phenotype
Non-coding DNA in front of the gene	Binding of RNA polymerase	Quantity of protein produced is altered
Coding DNA of the gene	Sequence of amino acids	Activity of the protein produced is altered

Quick check

- 1 . What decides the order of amino acids in a protein?
- 2 . What does RNA polymerase do?
- 3 . What is a codon?
- 4 . What carries amino acids to the ribosome?
- 5 . How can a variant in non-coding DNA affect phenotype?

4 Inheritance Spec 3.11B-3.18B

Gregor Mendel Spec 3.11B Triple only

Gregor Mendel was a monk who worked out the basis of genetics by breeding pea plants in the mid-1800s. He chose characteristics with clear alternatives, such as tall or short plants, and purple or white flowers.

- He crossed plants with different characteristics and counted the characteristics of the offspring over several generations.
- He found that characteristics are passed on as separate **inherited factors**, one from each parent. We now call these factors **genes**.
- He showed that some factors are **dominant** and others are **recessive**. For example, crossing pure-breeding tall plants with pure-breeding short plants gave all tall plants. Crossing those offspring with each other gave about three tall plants for every one short plant.

Difficulties before the mechanism was discovered

- Nobody knew about **genes, DNA or chromosomes**, so there was no mechanism to explain how characteristics are passed on.
- The behaviour of chromosomes during cell division and fertilisation had not been seen, so his ideas could not be linked to anything observable.
- His work was not recognised or understood at the time. It was only appreciated after the structures and processes involved in inheritance were discovered.

Exam tip: Link both halves: Mendel found patterns in how characteristics are inherited, but could not explain them because genes and chromosomes had not yet been discovered.

Alleles and inherited differences Spec 3.12

Different forms of the same gene are called **alleles**. A gene for flower colour may have an allele for purple and an allele for white.

In body cells chromosomes are in pairs, so an organism usually has two alleles of each gene: one inherited from each parent. Individuals can have different combinations of alleles, so they have **different inherited characteristics**.

The alleles of a gene have slightly different base sequences, so they can code for slightly different versions of the same protein. This is why a difference in alleles can cause a difference in a characteristic.

Common mistake: An allele is not a different gene. It is a different version of the same gene, found at the same position on a chromosome.

Key genetic terms Spec 3.13

Chromosome	A long molecule of DNA in the nucleus that carries many genes.
Gene	A section of DNA that codes for a specific protein.
Allele	A different version of the same gene.
Dominant	An allele that is expressed in the phenotype even when only one copy is present.
Recessive	An allele that is only expressed in the phenotype when two copies are present (no dominant allele).
Homozygous	Having two identical alleles of a gene, for example TT or tt.
Heterozygous	Having two different alleles of a gene, for example Tt.
Genotype	The alleles an organism has for a gene (or genes).
Phenotype	The characteristics that are expressed and can be observed.
Gamete	A sex cell (such as a sperm or egg) with half the number of chromosomes.
Zygote	The cell formed when two gametes fuse at fertilisation.

Dominant alleles are written as capital letters (T) and recessive alleles as lower case (t). A heterozygous plant, Tt, shows the dominant characteristic.

Exam tip: Genotype is the alleles, such as Tt. Phenotype is what you can see, such as tall. Do not swap them.

Monohybrid inheritance Spec 3.14

Monohybrid inheritance is the inheritance of a characteristic controlled by a single gene. You can show it using a genetic diagram, a Punnett square or a family pedigree.

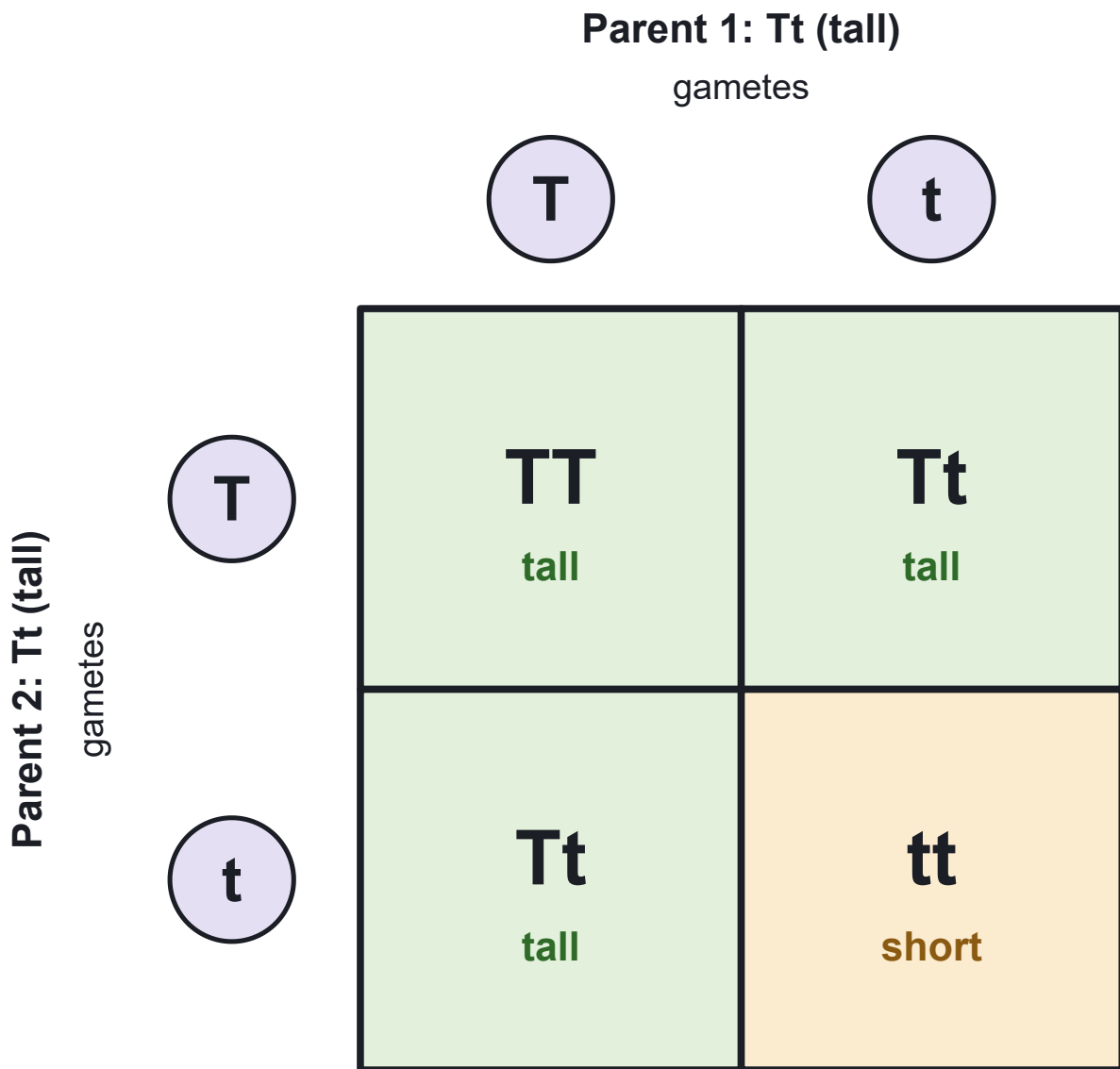
1. Choose letters for the alleles: a capital for dominant, lower case for recessive.
2. Write the genotypes of the parents.
3. Write the possible gametes from each parent. Each gamete has one allele.
4. Combine the gametes in a Punnett square to show the possible genotypes of the offspring.
5. Write the phenotype for each genotype.

Crossing two heterozygous pea plants

Tall (T) is dominant to short (t). Two heterozygous tall plants (Tt) are crossed. Show the possible offspring.

1. Parent genotypes: Tt and Tt.
2. Gametes: T or t from each parent.
3. Punnett square: top row (gametes from parent 1) T and t; side column (gametes from parent 2) T and t. The four boxes are TT, Tt, Tt and tt.
4. Phenotypes: TT, Tt and Tt are tall; tt is short.

Answer: Offspring genotypes: 1 TT : 2 Tt : 1 tt. Phenotypes: 3 tall : 1 short.

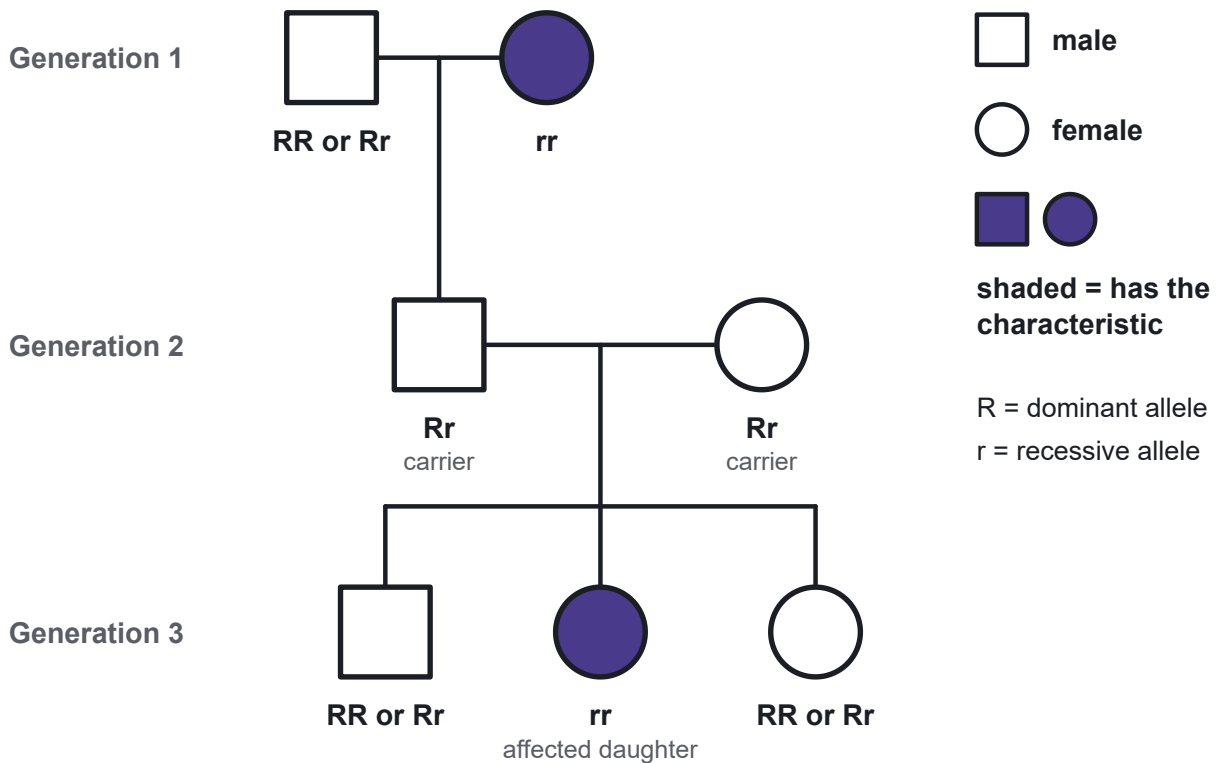


Tt × Tt gives 3 tall : 1 short (1 TT : 2 Tt : 1 tt).

Family pedigrees

A **family pedigree** (family tree) shows who in a family has a characteristic. Squares are usually males and circles are females. Shaded shapes show people with the characteristic. A horizontal line joins the parents and a vertical line leads down to their children.

- If two parents who do not have a characteristic have a child who does, the characteristic is **recessive**. Both parents must be **carriers**, heterozygous with one copy of the recessive allele.
- If two parents who both have a characteristic have a child who does not, the characteristic is **dominant**. Both parents must be heterozygous.



Two unaffected parents with an affected daughter: the characteristic is **recessive** and both parents are carriers (Rr).

Exam tip: In a pedigree question, start by finding two parents with the same phenotype and a child with the other phenotype. That tells you which allele is dominant.

Sex determination Spec 3.15

Humans have 23 pairs of chromosomes. One pair is the **sex chromosomes**. Females have two X chromosomes (XX). Males have one X and one Y chromosome (XY).

All eggs carry an X chromosome. Half of the sperm carry an X and half carry a Y. The sex of the offspring is decided at **fertilisation** by which sperm fertilises the egg.

Punnett square for sex determination		
	X (egg)	X (egg)
X (sperm)	XX female	XX female
Y (sperm)	XY male	XY male

Two outcomes are female and two are male, so the ratio is **1:1** and there is a **50% probability** that a baby will be a boy or a girl.

Common mistake: The father's sperm decides the sex of the baby, because it carries either an X or a Y. Every egg carries an X.

Calculating and analysing outcomes Spec 3.16

You need to calculate and analyse the outcomes of monohybrid crosses and pedigrees for dominant and recessive traits. Outcomes can be given as a ratio, a **probability** or a **percentage**.

Ways of giving the same outcome ($Tt \times Tt$)

Outcome	Boxes out of 4	Probability	Percentage
Short plant (tt)	1	1/4 or 0.25	25%
Tall plant (TT or Tt)	3	3/4 or 0.75	75%

As a **ratio**, this is 3 tall : 1 short. A 3 : 1 ratio means 1 in 4 offspring are short, not 1 in 3.

Probability for a recessive disorder

A disorder is caused by a recessive allele (f). Two parents do not have the disorder but both are carriers (Ff). What is the probability that their child has the disorder?

1. Gametes from each parent: F or f.
2. Possible offspring: FF, Ff, Ff and ff.
3. Only ff has the disorder. That is 1 out of 4 equally likely outcomes.

Answer: Probability = $1/4 = 0.25 = 25\%$.

Each child is an independent event. If the first child has the disorder, the probability for the next child is still 1 in 4. Probabilities describe chance. They do not guarantee the actual numbers in a family.

Maths skill: Show the Punnett square, count the favourable boxes out of the total, then convert to a ratio, fraction or percentage as the question asks.

ABO blood groups Spec 3.17B Triple only

The ABO blood group is controlled by a single gene with **multiple alleles**: more than two alleles of the same gene. There are three alleles, written here as I^A , I^B and I^O . A person inherits two, one from each parent.

Exam tip: In exam papers the second letter is printed small and raised above the I, but it means the same as I^A , I^B and I^O here. Some books write I^O as i .

I^A and I^B are **codominant**: if both are present, both are expressed in the phenotype. I^O is recessive to both.

ABO blood groups

Phenotype (blood group)	Possible genotypes
A	$I^A I^A$ or $I^A I^O$
B	$I^B I^B$ or $I^B I^O$
AB	$I^A I^B$
O	$I^O I^O$

Parents with group A and group B

A mother is $I^A I^O$ and a father is $I^B I^O$. What blood groups could their children have?

1. Mother's gametes: I^A or I^O . Father's gametes: I^B or I^O .
2. Possible children: $I^A I^B$, $I^A I^O$, $I^B I^O$ and $I^O I^O$.

Answer: Group AB, A, B and O, each with a 1 in 4 (25%) probability.

Exam tip: Define codominance as both alleles being expressed in the phenotype of a heterozygous person. Do not call it blending.

Sex-linked genetic disorders Spec 3.18B Triple only Higher tier

A **sex-linked** genetic disorder is caused by an allele on a sex chromosome. Most are caused by a recessive allele on the X chromosome. Haemophilia is an example.

- Males (XY) have only **one X chromosome**, so one copy of the recessive allele is enough to cause the disorder. The Y chromosome does not carry a matching allele.
- Females (XX) have two X chromosomes. They need **two copies** of the recessive allele to have the disorder. With one copy they are **carriers**.
- This is why sex-linked disorders are more common in males.

Carrier mother (XHXh) and unaffected father (XHY)

	XH (egg)	Xh (egg)
XH (sperm)	XHXH unaffected female	XHXh carrier female
Y (sperm)	XHY unaffected male	XhY affected male

Here XH is an X chromosome carrying the dominant normal allele (H) and Xh is an X chromosome carrying the recessive allele for haemophilia (h). The probability that a son has haemophilia is 50%. No daughters are affected, but half are carriers. An affected father passes his X to every daughter and his Y to every son, so his sons do not inherit the disorder from him.

Exam tip: Write the allele next to the X chromosome, for example XhY (exam papers print the h small and raised). Show the Y with no allele.

Quick check

1. What is an allele?
2. What is the difference between genotype and phenotype?
3. Which sex chromosomes do human females and males have?
4. Two parents without a disorder have an affected child. Is the allele dominant or recessive?
5. What is codominance?

5 Variation, mutations and the Human Genome Project Spec 3.19-3.23

Features controlled by multiple genes Spec 3.19

Some characteristics, like flower colour in Mendel's pea plants, are controlled by a single gene. These are the ones used in monohybrid crosses.

Most phenotypic features are the result of **multiple genes** rather than single gene inheritance. Several genes work together to produce the feature. Features controlled by many genes, such as height, show a wide range of values rather than a few clear categories.

Exam tip: If a question asks why a feature does not follow a simple 3 : 1 ratio, one answer is that it is controlled by more than one gene.

Causes of variation Spec 3.20

Differences between individuals of the same species are called **variation**. Variation in phenotype has two causes: genes and the environment. Often it is a combination of both.

Genetic and environmental variation		
	Genetic variation	Environmental variation
What it is	Different characteristics as a result of mutation and sexual reproduction	Different characteristics caused by an organism's environment (acquired characteristics)
Source	Differences in the alleles inherited	Conditions during life, such as diet, climate, injury or activity
Examples	Eye colour, sex	A scar, body mass affected by diet, plant height affected by light

Continuous and discontinuous variation

- Continuous variation**: the feature can take any value in a range, such as height or mass. The results are shown on a histogram.
- Discontinuous variation**: the feature falls into distinct categories with nothing in between, such as sex or purple versus white flowers. The results are shown on a bar chart.

Exam tip: Practical to know: investigate variation within a species, for example the length of leaves from one type of plant, or the height of students in a class. Measure a large sample, then draw a graph to see whether the variation is continuous or discontinuous.

Calculating an arithmetic mean

The heights of five seedlings grown in the same conditions were 12 cm, 15 cm, 11 cm, 14 cm and 13 cm. Calculate the mean height.

1. Add the values: $12 + 15 + 11 + 14 + 13 = 65$ cm.
2. Divide by the number of values: $65 \div 5 = 13$ cm.

Answer: Mean height = 13 cm.

Maths skill: The mean is the total of all the values divided by the number of values. Give the unit in your answer.

Mutations and genetic variation Spec 3.22, 3.23

A **mutation** is a change in the base sequence of DNA. There is usually **extensive genetic variation** within a population of a species, and this variation arises through mutations.

Mutations affect the phenotype in different ways:

- Most genetic mutations have **no effect** on the phenotype.
- Some mutations have a **small effect** on the phenotype.
- Rarely, a single mutation will **significantly affect** the phenotype.

Common mistake: Do not write that all mutations are harmful. Most have no effect on the phenotype.

The Human Genome Project Spec 3.21

The **Human Genome Project** was an international research project to map the human genome. It was completed in 2003 and identified the sequence of bases in human DNA and where the genes are.

Potential applications in medicine

- Finding the genes linked to inherited disorders, so that people at risk can be tested and their risk predicted.
- Developing new treatments and drugs, and tailoring treatment to a person's own genes.
- Understanding how genes are involved in disease, which could help prevent or diagnose it earlier.

Concerns

- People may not want to know if they are likely to develop a disease that cannot be cured, which could cause worry.
- Genetic information must be kept private. There are concerns that it could be used unfairly, for example by insurers or employers.

Exam tip: A 'discuss' question wants a balanced answer. Give at least one benefit and one concern, and link each to the genome information.

Quick check

1. State where genetic variation in a population comes from.
2. What is environmental variation?
3. What effect do most mutations have on the phenotype?
4. Give one possible medical use of the Human Genome Project.

Quick check answers

Reproduction and meiosis

1. Advantage: no need to find a mate (or rapid reproductive cycle). Disadvantage: no variation in the population.
2. Advantage: variation in the population. Disadvantage: the requirement to find a mate.
3. Four.
4. 23, which is half the number in a body cell (46).

DNA and the genome

1. A double helix.
2. A with T, and C with G.
3. A section of a DNA molecule that codes for a specific protein.
4. It breaks down the cell membranes to release the DNA.

Protein synthesis and gene variants

1. The order of bases in a section of DNA (the gene).
2. It binds to non-coding DNA in front of a gene and produces a complementary mRNA strand from the coding DNA.
3. A triplet of bases in mRNA that codes for a specific amino acid.
4. tRNA.
5. It can change the binding of RNA polymerase and so alter the quantity of protein produced.

Inheritance

1. A different version of the same gene.
2. Genotype is the alleles an organism has. Phenotype is the observable characteristic that is expressed.
3. Females XX, males XY.
4. Recessive, and both parents are carriers.
5. Both alleles are expressed in the phenotype when both are present, as with IA and IB giving blood group AB.

Variation, mutations and the Human Genome Project

1. Mutations (and sexual reproduction).
2. Different characteristics caused by an organism's environment (acquired characteristics).
3. No effect.
4. For example, identifying genes linked to inherited disorders so that people can be tested, or developing treatments.