

Genetics revision notes

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

For Edexcel GCSE Combined Science 1SC0, **Higher tier** is only on the Higher paper.

1 Reproduction and meiosis Spec 3.1B-3.3

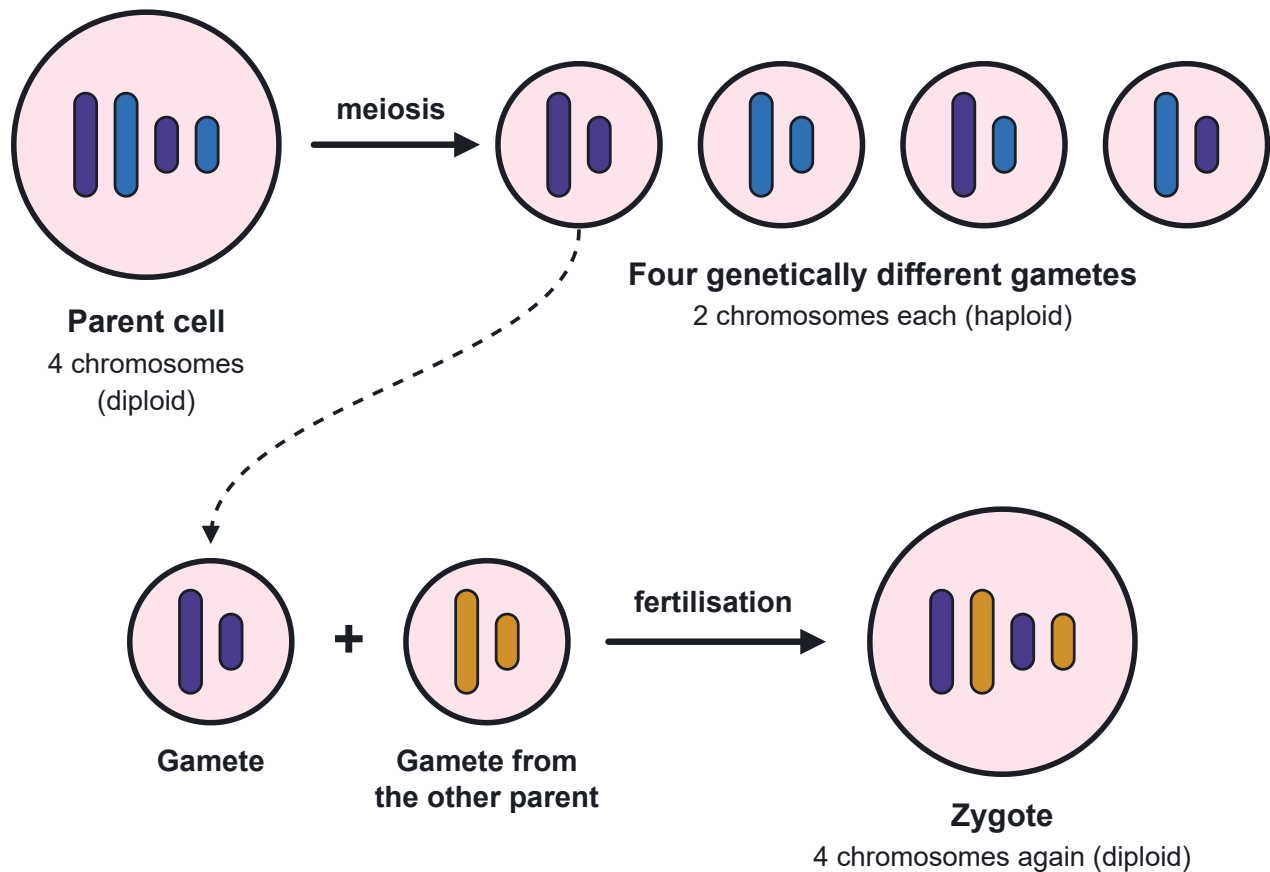
Meiosis Spec 3.3

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



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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

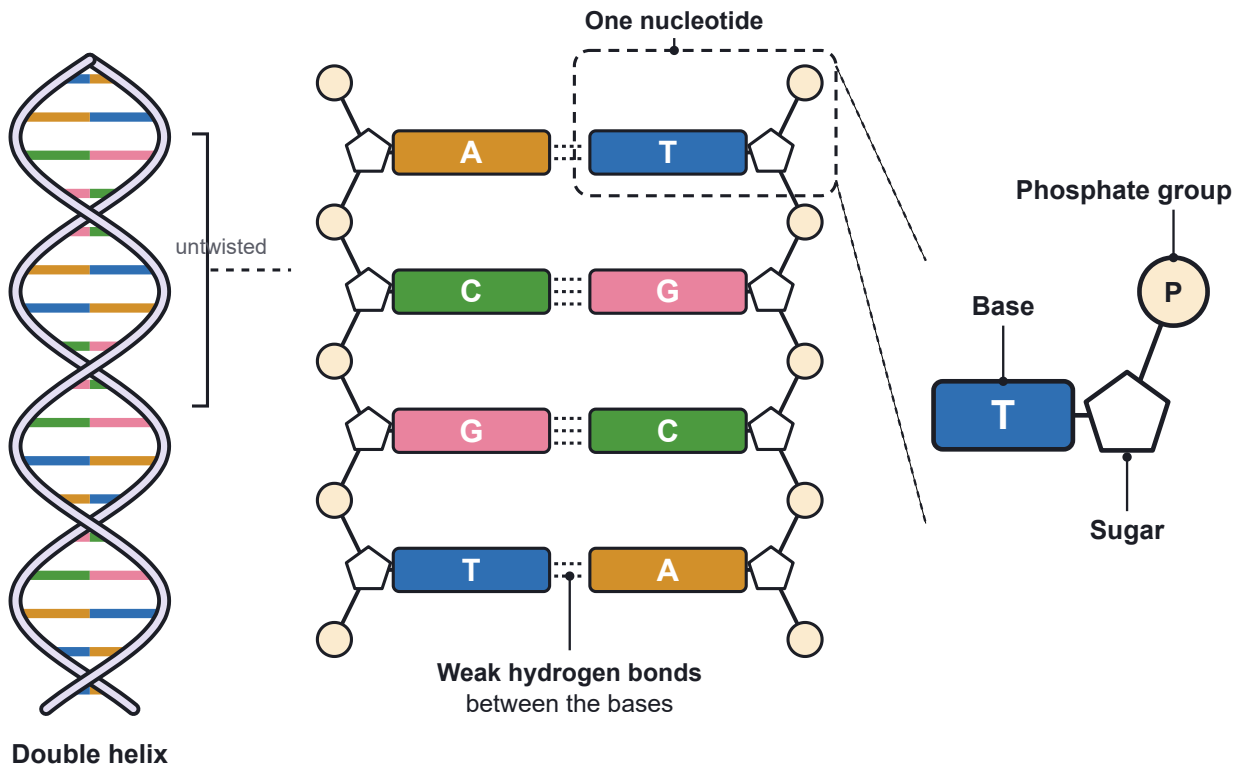
DNA is a **polymer** made up of many repeating units called **nucleotides**. A polymer is a long molecule made of many similar smaller units joined together.

- 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.



Complementary base pairs: A with T, C with G

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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 Inheritance Spec 3.11B-3.18B

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.

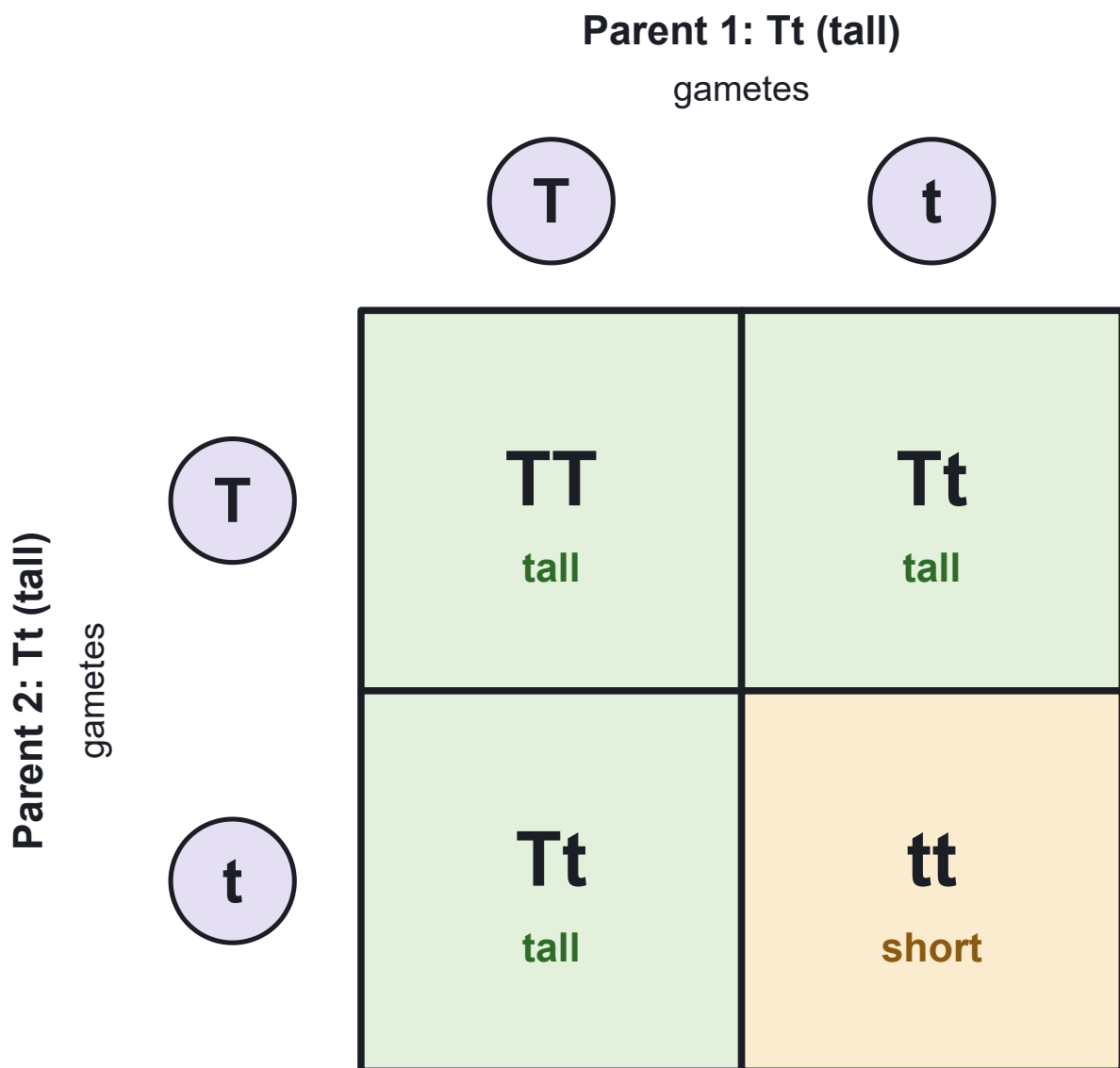
3. Write the possible gametes from each parent. Each gamete has one allele.
4. Combine the gametes in a Punnett square to show the possible genotypes of the offspring.
5. Write the phenotype for each genotype.

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.



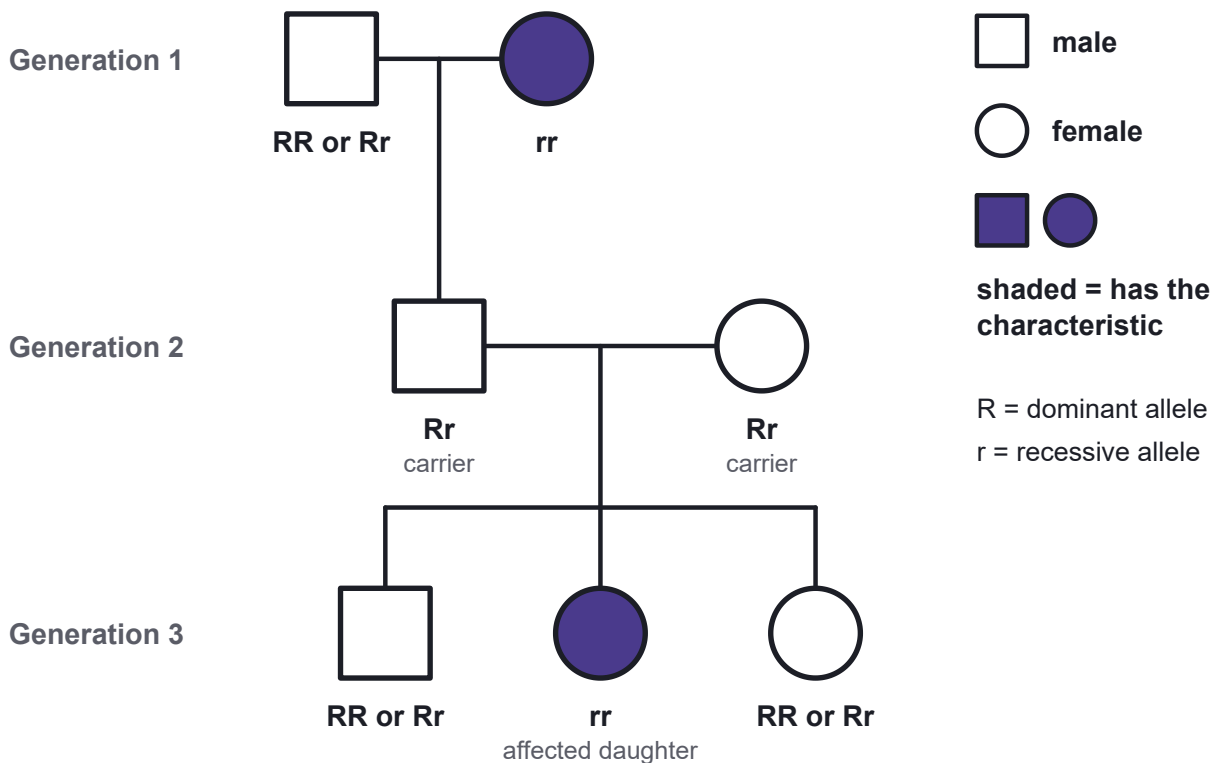
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Tt × Tt gives **3 tall : 1 short** (1 TT : 2 Tt : 1 tt).

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.



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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 × 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.

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?

4 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.

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.