

Variation, mutations and the Human Genome Project

Revision notes · Causes of variation, mutations and their effects, characteristics controlled by many genes, and the Human Genome Project.

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

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

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.