DNA and the genome

The whole genetic material of an organism is called its genome. You need to know about how genes work and the importance of the Human Genome Project.

10 Genes and the genome

The whole genetic material of an organism is called its **genome**.

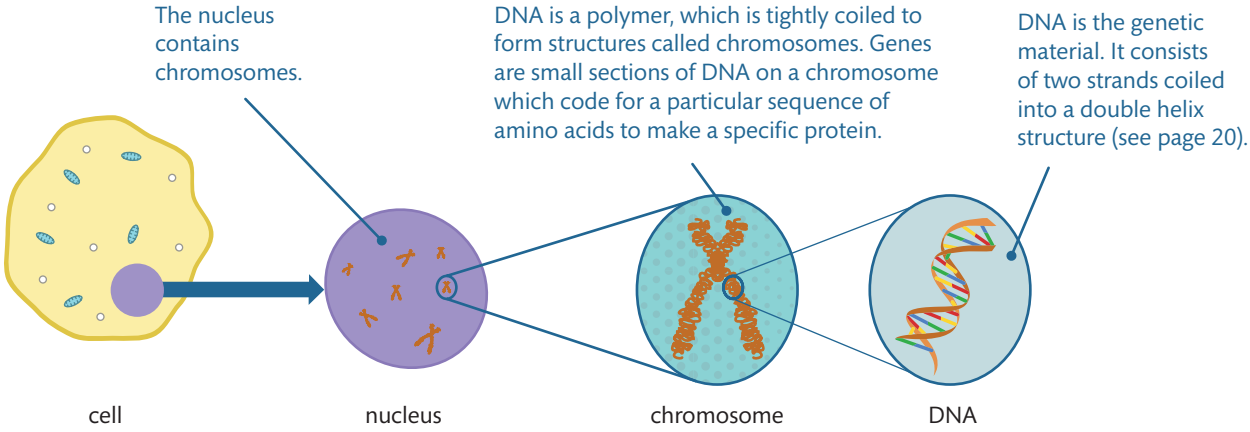


Figure 1 DNA in a cell

5 The Human Genome Project

Completed by 2003, the purpose of the **Human Genome Project** was to map and identify all the genes in the human genome.

Information about DNA can be very useful for forensic science, tracing human migration patterns, and for the understanding and treatment of inherited genetic disorders. The project helps scientists to:

- diagnose diseases before symptoms develop
- identify the genetic changes that are responsible for an already diagnosed disease
- help doctors to determine the best treatment
- identify genetic mutations that may increase the risk of developing a disease
- identify gene changes that could be inherited
- screen babies for treatable conditions.

5 Worked example — Grade 6–7

1 Describe the role of a gene. **[1 mark]**

A gene codes for a sequence of amino acids to make a protein.

2 Explain how the human genome and genetic markers have enabled scientists to trace human migration back to Africa. **[3 marks]**

Genetic markers are areas of variation in the sequence of human DNA. Mapping the occurrence of these markers around the world shows the movement of human population. As human populations migrated, their genome mutated, showing small changes in different populations. These changes can be traced back, showing the patterns of migration of human populations from their ancestors in Africa.

Virtually all human DNA is identical – only around 1 in 1000 DNA base pairs varies from one individual to the next. These variations are known as genetic markers, caused by mutations in DNA.

10 Exam-style practice — Grade 7

1 Cystic fibrosis is an inherited disorder. Explain how the human genome can be used to identify whether or not an unborn baby carries the disorder. **[3 marks]**

2 Describe how an understanding of the human genome is useful for developing treatments for diseases caused by mutations in genes. **[2 marks]**

3 Give **three** ways in which knowledge of the sequence of the human genome can be used. **[3 marks]**

Genetic inheritance

Many of our characteristics are controlled by the genes we inherit. You need to know how alleles, the different forms of each gene, cause variation between individuals.

5 Key terms

- **gene** – a short section of DNA which codes for a protein
- **allele** – different version of a gene
- **dominant** – only one dominant allele is needed for a characteristic to be expressed
- **recessive** – two recessive alleles are needed for the characteristic to be expressed
- **homozygous** – both alleles for a gene are identical
- **heterozygous** – the alleles for a gene are different
- **genotype** – the alleles present for genes
- **phenotype** – the physical characteristics, determined by the alleles

5 Genes

Some characteristics are controlled by a single **gene**, such as red-green colour blindness in humans, and fur colour in mice.

Most characteristics are controlled by multiple genes interacting. For example, multiple genes affect eye colour and skin colour.

The different forms of a gene are called **alleles**; alleles for each gene are inherited from each parent.

The combination of alleles present (**genotype**) operates at a molecular level to develop a person's observable characteristics (**phenotype**).

The appearance of a characteristic is dependent on both the type of alleles present and whether they are **dominant** or **recessive**.

10 Worked example — Grade 6

The allele **m** causes a rare blood-linked condition.

(a) What type of allele is responsible for the disease? **[1 mark]**

A recessive allele

(b) Two parents produce offspring. Parent A has the alleles **Mm** and parent B has the alleles **mm**.

Produce a Punnett square to calculate the percentage chance of their offspring inheriting the disease. Draw a circle around any offspring who have the disease. **[4 marks]**

	Parent A: M	Parent A: m
Parent B: m	Mm	mm
Parent B: m	Mm	mm

There is a 50% chance of having a child with the disease.

One mark is awarded for correctly identifying the alleles. The second mark is for four correct crosses. The third mark is for identifying the offspring who have the disease. The final mark is for the correct percentage given.

Dominant alleles are represented by a capital letter, while recessive alleles are shown by a lower case letter.

2 Working scientifically

You need to know how to draw genetic cross diagrams, such as **Punnett squares**, to predict the probability of the results of a single gene cross. (A different type of genetic diagram is shown on page 24.) The results of genetic crosses are usually represented as either a ratio or a percentage.

2 Exam focus

If a question asks you to draw a 'genetic diagram' then it is up to you which type you draw. However, a Punnett square is usually the easiest type to draw.

Go to page 23 to read about the inheritance of polydactyly.

5 Exam-style practice — Grade 6

Polydactyly is an inherited condition which causes a person to have extra fingers or toes. It is caused by a dominant allele.

(a) Draw a Punnett square to show the inheritance of polydactyly between a mother who is homozygous for polydactyly, and a father who does not have the polydactyly allele.

Use **D** for the polydactyly allele and **d** for the normal allele. **[3 marks]**

(b) Give the percentage of the offspring that will inherit the condition. **[1 mark]**

Inherited disorders

You need to know how genetic diagrams, such as Punnett squares and family pedigrees, show how genetic disorders are inherited.

5 Family pedigree diagrams

Family pedigree diagrams are family trees showing how inherited disorders are passed down through different generations. The example in **Figure 1** shows the inheritance of polydactyly.

Polydactyly is a disorder where the person has extra fingers or toes. It is caused by a dominant allele.

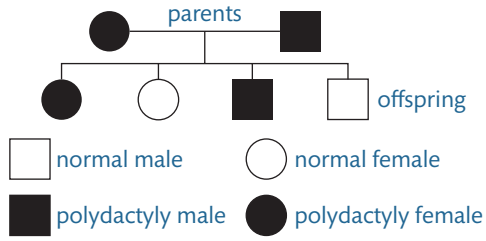


Figure 1 This family pedigree diagram shows the inheritance of polydactyly within a family.

1 Exam focus

In the exam, you could be asked to:

- complete a Punnett square diagram
- extract and interpret information from genetic cross and family pedigree diagrams.

2 Maths skills

When explaining the results of genetic diagrams, you may be asked to give your answers as:

- percentages (%)
- probabilities
- ratios.

A chance of 100% is the same as a probability of 1.0, and a chance of 50% is the same as a probability of 0.5.

Always give ratios in the lowest form. For example, if the ratio is 2:2, then write this as 1:1.

10 Worked example — Grade 8

1 Polydactyly is an inherited disorder.

(a) Explain what is meant by 'an inherited disorder'. **[1 mark]**

An inherited disorder is a genetic disorder that has been passed on from either one or both parents.

(b) Explain how **Figure 1** shows that polydactyly is caused by a dominant allele and not by a recessive allele. **[4 marks]**

Two of the offspring do not have polydactyly, but both of the parents do. This means that polydactyly is caused by a dominant allele and each of the parents must be heterozygous. The offspring without polydactyly must have inherited a normal recessive allele from each parent.

If polydactyly was caused by a recessive allele, then both parents would have to be homozygous recessive and therefore all the offspring would have polydactyly too.

2 Draw and complete a Punnett square to show how two people with polydactyly can have a child without polydactyly. Use **D** for the allele causing polydactyly and **d** for the recessive allele. Identify any children without polydactyly by drawing a circle around them. **[3 marks]**

	D	d
D	DD	Dd
d	Dd	(dd)

10 Exam-style practice — Grade 7

Cystic fibrosis is a disorder of cell membranes leading to the production of a thick sticky mucus which affects organs, particularly the lungs. It is caused by a recessive allele.

The allele for cystic fibrosis can be represented by **f**, while the allele for the normal gene is represented as **F**.

(a) Give the genotype of a person who is unaffected and is not a carrier of the condition. **[1 mark]**

(b) Explain whether a person who has cystic fibrosis will be heterozygous or homozygous for the condition. **[2 marks]**

(c) Determine the probability of a child being affected if both parents are unaffected carriers. Use a Punnett square to show your answer. **[3 marks]**

Sex determination

You need to know how sex is determined by chromosomes.

Sex chromosomes

Human body cells each contain 23 pairs of chromosomes, 22 of which control characteristics that do not depend on whether you are male or female.

The 23rd pair carries the genes that determine whether a person is male or female.

The female sex chromosomes are XX. The male sex chromosomes are XY.

Sex cells (gametes) only contain 23 chromosomes, one of each pair. Female sex cells (gametes) therefore only contain one X chromosome. The male sex cells (gametes) can either contain one X or one Y, depending on how the chromosomes are separated during meiosis. Therefore an X chromosome is always inherited from the egg but there is a 50% chance of inheriting either an X or a Y chromosome from the sperm, which determines the sex of the offspring.

Genetic diagrams

A genetic diagram can be used to determine the sex of offspring.

1. To construct a genetic diagram, the phenotype of each parent must be given on the top line.
2. The next stage is to provide the genotype for each parent, underneath their phenotype.
3. The next line shows the genotype of all the gametes which can be passed on to the offspring.
4. The final stage in the genetic cross diagram is to show all of the combinations of gametes which could occur which gives the different genotypes possible.

You are expected to know the genotype for a male and a female:

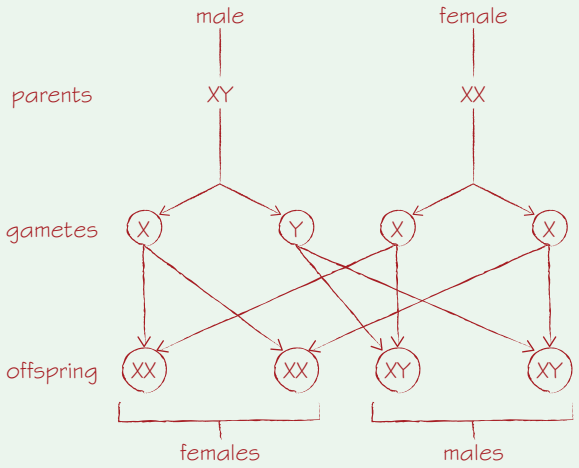
- In males, the two sex chromosomes are different. They are XY.
- In females, the two sex chromosomes are the same. They are XX.

Working scientifically

Although it is possible to identify the sex of an unborn baby, it is illegal to choose the sex of a baby unless you have a serious genetic condition which could put a child at risk if inherited, such as haemophilia or muscular dystrophy. (The alleles for these conditions are carried on the sex chromosomes.)

Worked example — Grade 7

There is a 1 : 1 chance of a child being a boy or a girl. Draw a suitable diagram to explain this. **[4 marks]**



The diagram shows that there is a 2 : 2 chance of female : male offspring, which equates to a 1 : 1 ratio.

You could also show your answer as a Punnett square diagram:

	gametes	Male: X	Male: Y
Female	X	XX	XY
	X	XX	XY

Exam-style practice — Grade 5

Humans have two different sex chromosomes: X and Y.

(a) Give the genotype of a female. **[1 mark]**

(b) Give the number of chromosomes, passed on from the egg, that help determine the sex of a baby. **[1 mark]**

(c) Give the probability of having a male child. Explain your answer. **[2 marks]**

Variation and mutation

You need to know how both variation and mutation occur in a species.

10 Causes of variation

Although a lot of characteristics are inherited, the interaction of genes with the environment will also influence the phenotype (physical features) of an individual (see page 22).

Variation between individuals occurs due to differences in:

- inherited genes
- the environment in which the person grew up
- the interaction of genes with the environment.

Some characteristics brought about by genetic variation include Down's syndrome, blood group and eye colour.

Some characteristics brought about by environmental variation include language spoken and the presence of scars. Characteristics caused by the environment are sometimes called **acquired characteristics**.

Most examples of variation are influenced by both environmental and genetic factors. For example, a person may have the potential to be tall, but an unhealthy diet can cause poor growth.

There is usually a lot of genetic variation within a population of a species.

Genetic variation in individuals is caused by sexual reproduction (see pages 15 and 19) and by **mutations**. Most mutations will have no effect on the phenotype; some mutations have a small effect on the phenotype; rarely, a single mutation will significantly affect the phenotype. Mutations occur continuously. If the new phenotype is suited to an environmental change it can lead to a relatively rapid change in the species.

Mutations can have positive or negative effects:

- improve chances of survival
- increase genetic diversity
- can lead to diseases, such as cancer (pages 15 and 41)
- can lead to genetic disorders (page 23).

5 Mutations: key facts

- A mutation is a change in genetic material (DNA).
- Mutations occur naturally and continuously, usually when DNA is being copied before cell division takes place.
- Mutations usually have no effect on an individual's characteristics. However, sometimes they can be harmful or useful.
- Mutations cause variation within a species, which can be vital to ensure the survival of the species.
- Some mutations are caused by substances such as tar from cigarettes.
- Radiation, including gamma rays, X-rays and UV rays, can also cause genetic mutations.

10 Worked example — Grades 4–6

Identical twins are individuals who developed from a single fertilised ovum (egg cell). As a result, they have identical DNA. Explain why identical twins may or may not be identical when they reach adulthood. **[3 marks]**

Phenotype is controlled by genetic inheritance, environmental factors and the interaction between them. Identical twins will inherit the same DNA and so are genetically identical. However, environmental factors such as their diet will also affect their phenotypes so their appearances could be different.

Remember, the phenotype (appearance) is determined by a combination of factors.

5 Exam-style practice — Grade 7

1. Give **two** examples of genetic variation and **two** examples of environmental variation. **[2 marks]**
2. Describe what is meant by a mutation. Give **two** causes of mutation. **[3 marks]**
3. Give **one** way in which a mutation can be beneficial. **[1 mark]**

Evolution by natural selection

You need to know how the theory of evolution explains the development of species over time.

5 Natural selection

The theory of **evolution** states that all species of living things have evolved, over more than three billion years, from simple life forms, through a process called natural selection. **Natural selection** is the theory that organisms which are best suited to their environment are more likely to survive and reproduce. Therefore, their offspring are more likely to inherit genes that give rise to phenotypes (see pages 22 and 25) most suited to the environment, causing changes to the population over time.

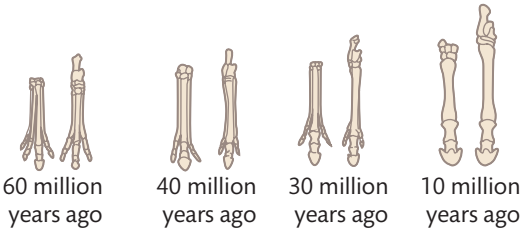


Figure 1 All the main stages of horse evolution have been preserved in fossil records.

5 Working scientifically

Alongside other scientists, **Charles Darwin** developed the theory of natural selection after observing many examples of variation, for example, between species of tortoises and finches, while voyaging around South America and the Galapagos Islands.

Darwin realised that the animals that exhibited a variation that made them more successful were more likely to survive and breed, passing on the genes (alleles) for this feature to their offspring. As later generations reproduce, the successful feature continues to be passed on to the next generation.

As more evidence surrounding genetic inheritance (page 22) has been discovered and more of the fossil record continues to be discovered, the theory of evolution by natural selection has become widely accepted by scientists. There is further evidence for evolution in the process by which bacteria become resistant to certain antibiotics over time (page 39), and in how some pests, such as rats or mosquitoes, become resistant to particular poisons or insecticides.

The theory of evolution by natural selection was only gradually accepted because:

- the theory challenged the idea that God made all the animals and plants that live on Earth
- there was insufficient evidence at the time the theory was first published to convince all scientists
- the mechanism of inheritance and variation was not known until 50 years after the theory was published.

10 Worked example — Grade 4

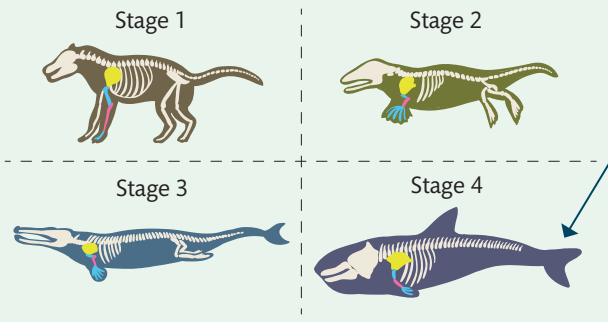


Figure 2 Evolution of the whale

Figure 2 shows how the whale is thought to have evolved from its land-dwelling ancestors.

Give **two** ways in which the whale has evolved since Stage 1. **[2 marks]**

The early ancestor had four limbs, and the whale alive today doesn't.

The whale has a fin on its back unlike its ancestors.

New species can form when the genes of individuals within a species become so different that their phenotype changes and they can no longer interbreed to form fertile offspring. This is known as **speciation**.

You are not expected to recall evolutionary stages in the development of species; you need to use the information provided in the question.

You could also be asked to explain why the feature has evolved. For example, the whale's fin developed so that the whale could swim efficiently.

5 Exam-style practice — Grade 7

1. Define the theory of evolution. **[2 marks]**
2. Some species have evolved to look like another species; for example, the scarlet king snake, which is harmless, has evolved to look like the deadly coral snake. Explain why this is considered to be a successful adaptation. **[2 marks]**
3. Describe the process of natural selection suggested by Darwin. **[4 marks]**