DNA and the genome

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10 Genes and the genome

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

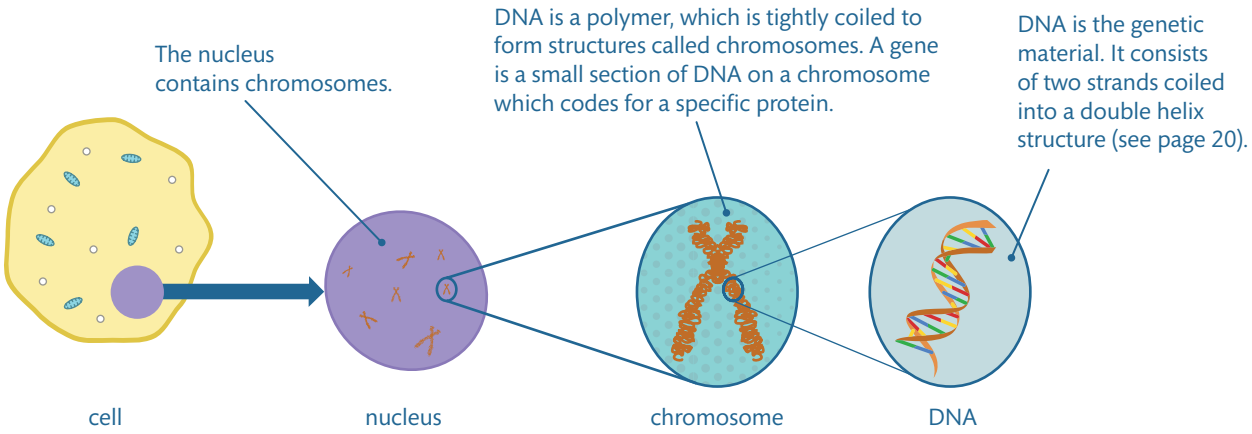


Figure 1 DNA in a cell

5 Worked example Grades 4–5

1 Explain the difference between a gene and a person's genome. **[2 marks]**

A gene is a section of DNA that codes for a particular protein. The genome is all the DNA or genes of a person.

2 Give **two** reasons why it could be useful to find out if a person is carrying a gene for a genetic disorder, even if they are showing no symptoms or signs of illness. **[2 marks]**

They can be prepared if symptoms develop in future. They can decide whether to have children who might inherit the genetic disorder.

3 The Human Genome Project took 13 years to complete. This was shorter than had originally been planned. Suggest **two** reasons why it was completed sooner than originally planned. **[2 marks]**

Many different scientists worked together on the project. Technology was developed that speeded up the process.

You could also have said that humans had fewer genes than scientists originally thought.

5 Working scientifically

Completed by 2003, the purpose of the **Human Genome Project** was to map and identify all the genes in the human genome. It was an international project involving scientists from 20 organisations, in six different countries.

Information about DNA can be very useful for forensic science, 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 Exam-style practice Grades 3–5

1 What does a gene code for? **[1 mark]**

- ☐ **A** carbohydrate
- ☐ **B** DNA
- ☐ **C** lipid
- ☐ **D** protein

2 Arrange the following in order of size, starting with the smallest. **[2 marks]**

cell	chromosome	gene	nucleus

3 Suggest how knowledge of DNA could help forensic scientists investigate a crime scene at which blood has been found. **[2 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 seen in the phenotype (expressed)
- **recessive** – two recessive alleles are needed for a characteristic to be seen in the phenotype (expressed)
- **homozygous** – both alleles for a gene are identical
- **heterozygous** – the alleles for a gene are different
- **genotype** – the alleles of a gene present in an individual
- **phenotype** – the physical characteristics, determined by the alleles

10 Worked example Grades 3–5

In mice, the allele for grey fur, **G**, is dominant to the allele for white fur, **g**.

(a) Give the phenotype of a mouse with the genotype **gg**. **[1 mark]**

White fur.

(b) Give **one** of the possible genotypes of a mouse with grey fur. **[1 mark]**

GG.

The other possible genotype is Gg.

(c) If the allele **G** is dominant, give the term that describes the allele **g**. **[1 mark]**

Recessive.

Dominant alleles are represented by capital letters, and recessive alleles are shown by lower case letters. In an exam, if you have a choice of the letters you use for alleles, choose letters where the upper and lower cases are easily told apart, for example, A and a. Do **not** choose letters such as O and o, which can look very similar when handwritten.

(d) Complete the Punnett square to show the result of a cross between two mice with grey fur. Include both the genotypes and the phenotypes of the offspring.
State the chance of the offspring having white fur. **[3 marks]**

		mother: G	mother: g
father	G	GG grey	Gg grey
	g	Gg grey	gg white

There is a 25% chance of the offspring having white fur.

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**; one allele for each gene is 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**.

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.
The results of genetic crosses are usually represented as either a ratio or a percentage.

Another type of genetic diagram is shown on page 24.

5 Exam-style practice Grades 2–3

Use words from the box to complete the sentence. **[2 marks]**

alleles chromosomes genes genotypic phenotypic

Some characteristics are controlled by single genes with several _____, but most _____ features are controlled by several genes, which can make it very difficult to predict the characteristics of offspring.

Genetic diagrams

You need to know how genetic diagrams, such as Punnett squares and family pedigrees, show how some characteristics, including 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.

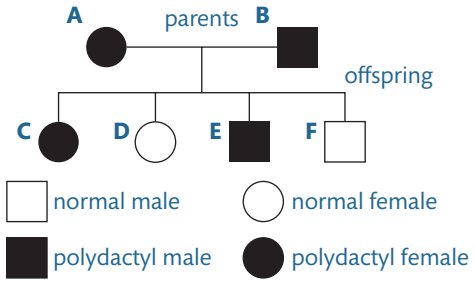


Figure 1 This family pedigree diagram shows the inheritance of polydactyly within a family

10 Worked example Grades 3–5

Look at **Figure 1**. Polydactyly is caused by a dominant allele.

(a) Give the sex for both **C** and **F**. For each person, also state whether or not they have polydactyly. **[2 marks]**

C is a female with polydactyly. F is a male who does not have polydactyly.

(b) Explain why both **A** and **B** have polydactyly, but have had some children who do **not** have polydactyly. **[2 marks]**

Polydactyly is caused by a dominant allele, so A and B must both be heterozygous and also carry a recessive allele. The children without polydactyly must have inherited two recessive alleles, one from each parent.

Exam focus

When writing out heterozygous genotypes, it is usual to put the capital letter first: e.g. write Bb rather than bB.

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.

2 Exam focus

If a question asks you to draw a 'genetic diagram', then it is up to you which type you draw. There are other types of genetic diagram, however, a Punnett square is usually the easiest to draw.

10 Exam-style practice Grades 3–5

The allele for brown eyes **B** is dominant to the allele for blue eyes **b**. Use this information to answer the following questions.

(a) Complete the Punnett square to show the possible offspring of a couple where one parent has blue eyes and the other parent has brown eyes but is heterozygous for eye colour.

Include both the genotypes and the phenotypes of the offspring.

State the probability of one of the children having blue eyes. **[3 marks]**

heterozygous brown eyes	blue eyes: b	blue eyes: b
B		
b		

(b) Explain whether two parents with blue eyes can have a child with brown eyes. **[2 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

On pages 22 and 23 you saw one type of genetic diagram called a Punnett square. In the **Worked example** you can see a different type of genetic diagram. In the example, the diagram is being used to determine the sex of offspring.

1. To construct this type of genetic diagram, the phenotype of each parent must be written on the top line.
2. The next stage is to write the genotype for each parent, underneath their phenotype.
3. The next line shows the genotypes of all the gametes which can be passed on to the offspring. These are usually drawn in circles.
4. The final stage in the genetic cross diagram is to show all of the combinations of gametes which could occur which give the different genotypes possible.

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

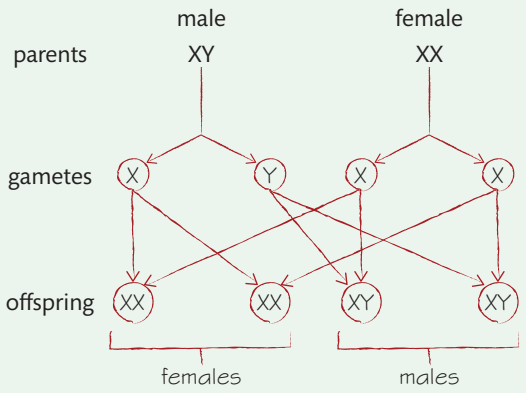
- In males, the two sex chromosomes are different. They are XY.
- In females, the two sex chromosomes are the same. They are XX.

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 5

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



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

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

		Male	
		X	Y
Female	X	XX	XY
	X	XX	XY

Exam-style practice — Grades 1–2

1 **(a)** What sex chromosomes are in a human egg cell? **[1 mark]**

☐ **A** X ☐ **B** Y ☐ **C** X or Y ☐ **D** X and Y

(b) What sex chromosomes are in a human sperm cell? **[1 mark]**

☐ **A** X ☐ **B** Y ☐ **C** X or Y ☐ **D** X and Y

2 Give the probability of a baby being a girl. **[1 mark]**

Variation and mutation

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

10 Causes of variation

The differences between different individuals' phenotypes are called **variation**.

Variation occurs due to differences in:

- inherited alleles
- the environment – this includes anything that happens to an individual during their lifetime
- the interaction between genes and the environment.

Some characteristics caused by genes include blood group and eye colour.

Some characteristics caused by the environment 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 genetic 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 genetic changes called **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 new species – this is evolution (see page 26).

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.
- The effect of a mutation on an individual's characteristics may be neutral, harmful or beneficial.
- 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.

5 Worked example Grades 2–4

1 Complete the table to show whether each example of variation is caused by genetic factors, environmental factors, or both. **[3 marks]**

Example	Genetic	Environmental	Both
blood group	✓		
body mass			✓
eye colour	✓		
hair length		✓	
height			✓
scars		✓	

2 Identical twins have the same DNA but sometimes can grow up to look quite different. Give **two** reasons why they might look different. **[2 marks]**

Live in different climates. Eat different foods.

Other possible answers include: different lifestyles, whether or not they smoke, different levels of activity.

5 Exam-style practice Grades 4–5

1 What is the best definition of a mutation? **[1 mark]**

- ☐ **A** change caused by growth
- ☐ **B** change caused by radiation
- ☐ **C** change in someone's DNA
- ☐ **D** change in the environment

2 Which of the following does **not** always cause variation between individuals? **[1 mark]**

- ☐ **A** ageing
- ☐ **B** environmental factors
- ☐ **C** mutation
- ☐ **D** sexual reproduction

Evolution by natural selection

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

5 Natural selection

The theory of **evolution** by natural selection states that all species of living things have evolved, over more than three billion years, from simpler 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.

The stages in natural selection are:

1 **Variation**: there are differences within a population – e.g. some giraffes are taller than others.

2 **Survival of the best adapted (sometimes called 'survival of the fittest')**: individuals with the best suited characteristics are more likely to survive – e.g. taller giraffes can reach more food in trees.

3 **Inheritance**: individuals with the best characteristics are more likely to breed and pass on their genes (alleles) to their offspring – e.g. taller giraffes are more likely to breed and have offspring who are tall.

4 **Change in species**: over many generations, genes (alleles) for the successful characteristics spread through the species – e.g. the average height of the giraffe population increases.

Figure 1 Taller giraffes have an advantage because they can reach leaves to eat that others cannot

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

You do not need to learn how particular species have evolved, and in the exam you will be given the information you need to answer the question.

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

5 Working scientifically

Charles Darwin developed the theory of natural selection after observing many examples of variation, for example, between different species of tortoise or of birds while voyaging around South America and the Galapagos Islands. Another scientist, Alfred Russel Wallace, also developed similar ideas.

As more evidence surrounding genetic inheritance (page 22) was discovered and more of the fossil record was found, the theory of evolution by natural selection became widely accepted. 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 not widely accepted at first 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
- how genes are inherited was not known until about 100 years after the theory was published.

5 Worked example — Grade 4

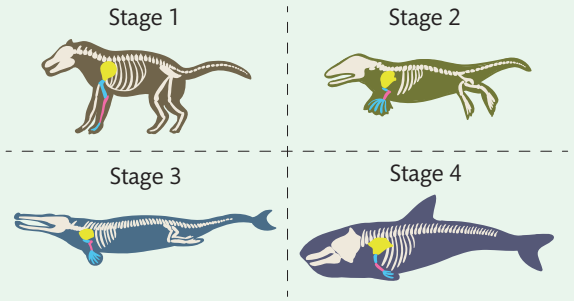


Figure 2

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.

10 Exam-style practice — Grades 3–5

1 Name the theory that Charles Darwin and Alfred Russel Wallace developed. **[1 mark]**

2 Cheetahs live in Africa and hunt antelope and other prey. Cheetahs are the fastest land animal in the world. Describe the stages of natural selection by which cheetahs evolved from slower-moving ancestors. **[4 marks]**