

B2 5.4

From Mendel to DNA

Learning objectives

- What did Mendel's experiments teach us about inheritance?
- What is DNA?
- How are specific proteins made in the body? [H]

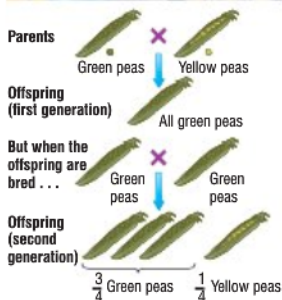


Figure 1 Gregor Mendel, the father of modern genetics. His work was not recognised in his lifetime but now we know just how right he was!

Until about 150 years ago people had no idea how information was passed from one generation to the next. Today we can identify people by the genetic information in their cells.

Mendel's discoveries

Gregor Mendel was born in 1822 in Austrian Silesia. He was clever but poor, so he became a monk to get an education.

He worked in the monastery gardens and became fascinated by the peas growing there. He carried out some breeding experiments using peas. He used smooth peas, wrinkled peas, green peas and yellow peas for his work. Mendel cross-bred the peas and counted the different offspring carefully. He found that characteristics were inherited in clear and predictable patterns.

Mendel explained his results by suggesting there were separate units of inherited material. He realised that some characteristics were dominant over others and that they never mixed together. This was an amazing idea for the time.

a Why did Gregor Mendel become a monk?

Mendel kept records of everything he did, and analysed his results. This was almost unheard of in those days. Eventually in 1866 Mendel published his findings.

He had never seen chromosomes nor heard of genes. Yet he explained some of the basic laws of genetics using mathematical models in ways that we still use today.

Mendel was ahead of his time. As no one knew about genes or chromosomes, people simply didn't understand his theories. He died 20 years later with his ideas still ignored – but convinced that he was right.

b What was unusual about Mendel's scientific technique at the time?

Sixteen years after Mendel's death, his work was finally recognised. By 1900, people had seen chromosomes through a microscope. Other scientists discovered Mendel's papers and repeated his experiments. When they published their results, they gave Mendel the credit for what they observed.

From then on ideas about genetics developed rapidly. It was suggested that Mendel's units of inheritance might be carried on the chromosomes seen under the microscope. And so the science of genetics as we know it today was born.

DNA – the molecule of inheritance

The work of Gregor Mendel was just the start of our understanding of inheritance. Today, we know that our features are inherited on genes carried on the chromosomes found in the nuclei of our cells.

These chromosomes are made up of long molecules of a chemical known as DNA (deoxyribonucleic acid). This has a double helix structure. Your genes are small sections of this DNA. The DNA carries the instructions to make the proteins that form most of your cell structures. These proteins also include the enzymes that control your cell chemistry. This is how the relationship

between the genes and the whole organism builds up. The genes make up the chromosomes in the nucleus of the cell. They control the proteins, which make up the different specialised cells that form tissues. These tissues then form organs and organ systems that make up the whole body.

Higher

The genetic code

The long strands of your DNA are made up of combinations of four different chemical bases (see Figure 2). These are grouped into threes and each group of three codes for an amino acid.

Each gene is made up of hundreds or thousands of these bases. The order of the bases controls the order in which the amino acids are put together so that they make a particular protein for use in your body cells. Each gene codes for a particular combination of amino acids, which make a specific protein.

A change or mutation in a single group of bases can be enough to change or disrupt the whole protein structure and the way it works.

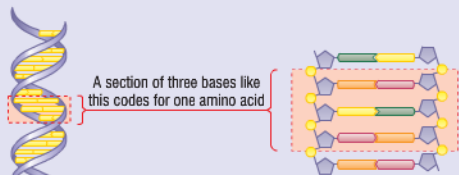


Figure 2 DNA codes for the amino acids that make up the proteins that make up the enzymes that make each individual

DNA fingerprinting

Unless you have an identical twin, your DNA is unique to you. Other members of your family will have strong similarities in their DNA. However, each individual has their own unique pattern. Only identical twins have the same DNA. That's because they have both developed from the same original cell.

The unique patterns in your DNA can be used to identify you. A technique known as 'DNA fingerprinting' can be applied to make the patterns known as **DNA fingerprints**.

These patterns are more similar between people who are related than between total strangers. They can be produced from very tiny samples of DNA from body fluids such as blood, saliva and semen.

The likelihood of two identical samples coming from different people (apart from identical twins) is millions to one. As a result, DNA fingerprinting is very useful in solving crimes. It can also be used to find the biological father of a child when there is doubt.

Did you know ... ?

The first time DNA fingerprinting was used to solve a crime, it identified Colin Pitchfork as the murderer of two teenage girls and cleared an innocent man of the same crimes.

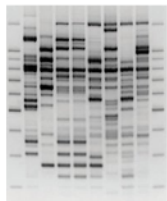


Figure 3 A DNA fingerprint

Summary questions

- How did Mendel's experiments with peas convince him that there were distinct 'units of inheritance' that were not blended together in offspring?
 - Why didn't people accept his ideas?
 - The development of the microscope played an important part in helping to convince people that Mendel was right. How?
- Two men claim to be the father of the same child. Explain how DNA fingerprinting could be used to find out which one is the real father.
- Explain the saying 'One gene, one protein'.

[H]

Key points

- Gregor Mendel was the first person to suggest separately inherited factors, which we now call genes.
- Chromosomes are made up of large molecules of DNA.
- A gene is a small section of DNA that codes for a particular combination of amino acids, which make a specific protein. [H]
- Everyone (except identical twins) has unique DNA that can be used to identify them using DNA fingerprinting.

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Inheritance in action

Learning objectives

- How is sex determined in humans?
- How do we predict what features a child might inherit?
- Can you construct a genetic diagram? [H]

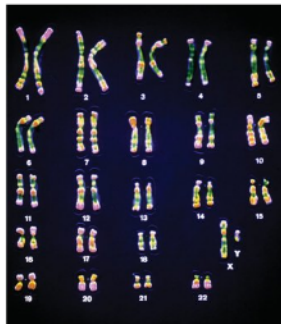


Figure 1 This special photo shows the 23 pairs of human chromosomes. You can see the XY chromosomes, which tell you they are from a male.

The way features are passed from one generation to another follows some clear patterns. We can use these to predict what may be passed on.

How inheritance works

Scientists have built on the work of Gregor Mendel. We now understand how genetic information is passed from parent to offspring.

Humans have 23 pairs of chromosomes. In 22 cases, each chromosome in the pair is a similar shape. Each one has genes carrying information about the same things. One pair of chromosomes is different – these are the **sex chromosomes**. Two X chromosomes mean you are female; one X chromosome and a much smaller one, known as the Y chromosome, mean you are male.

a Twins are born. Twin A is XY and twin B is XX. What sex are the two babies?

The chromosomes we inherit carry our genetic information in the form of genes. Many of these genes have different forms or alleles. Each allele will result in a different protein.

Picture a gene as a position on a chromosome. An allele is the particular form of information in that position on an individual chromosome. For example, the gene for dimples may have the dimple (D) or the no-dimple (d) allele in place.

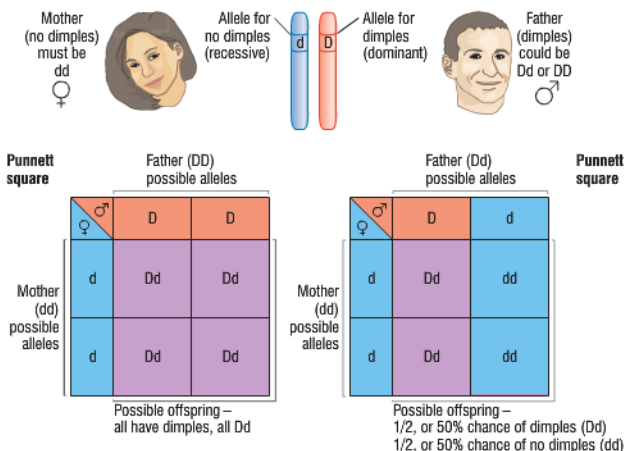


Figure 2 The different forms of genes, known as alleles, can result in the development of quite different characteristics. Genetic diagrams like these Punnett squares help you explain what is happening and predict what the offspring might be like.

Most of your characteristics, like your eye colour and nose shape, are controlled by a number of genes. However, some characteristics, like dimples or having attached earlobes, are controlled by a single gene. Often there are only two possible alleles for a particular feature. However, sometimes you can inherit one from a number of different possibilities. We can use biological models like the Punnett square in Figure 2 to predict the outcome of different genetic crosses.

Some alleles control the development of a characteristic even when they are only present on one of your chromosomes. These alleles are **dominant**, e.g. dimples and dangly earlobes. We use a capital letter to represent them, e.g. D.

Some alleles only control the development of a characteristic if they are present on both chromosomes – in other words, no dominant allele is present. These alleles are **recessive**, e.g. no dimples and attached earlobes. We use a lower case letter to represent them, e.g. d.

Higher

Genetic terms

Some words are useful when you are working with biological models such as Punnett squares or family trees:

- **Homozygous** – an individual with two identical alleles for a characteristic, e.g. DD, dd.
- **Heterozygous** – an individual with different alleles for a characteristic, e.g. Dd.
- **Genotype** – this describes the genetic makeup of an individual regarding a particular characteristic, e.g. Dd, dd.
- **Phenotype** – this describes the physical appearance of an individual regarding a particular characteristic, e.g. dimples, no dimples.

Family trees

You can trace genetic characteristics through a family by drawing a family tree. Family trees show males and females and can be useful for tracing family likenesses. They can also be used for tracking inherited diseases, showing a physical characteristic or showing the different alleles people have inherited.

Examiner's tip

When you choose a letter as a genetic symbol, try and use a letter that looks different in upper and lower case. Whatever you choose, be very careful to make the upper and lower case symbols clear. [H]

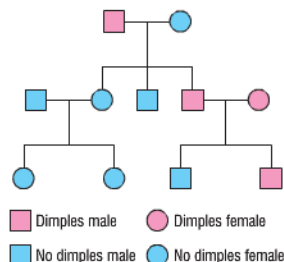


Figure 3 A family tree to show the inheritance of dimples

Summary questions

- 1 Copy and complete using the words below:
male sex chromosomes 23 22 X XX Y
Humans have pairs of chromosomes. In pairs the chromosomes are always the same. The final pair are known as
If you inherit you will be female, while an and a chromosome make you
- 2 a What is meant by the term 'dominant allele'?
b What is meant by the term 'recessive allele'?
c Try and discover as many human characteristics as you can that are inherited on a single gene. Which alleles are dominant and which are recessive?
- 3 Draw a Punnett square like the ones in Figure 2 to show the possible offspring from a cross between two people who both have dimples and the genotype Dd. [H]

Key points

- In human body cells the sex chromosomes determine whether you are female (XX) or male (XY).
- Some features are controlled by a single gene.
- Genes can have different forms called alleles.
- Some alleles are dominant and some are recessive.
- We can construct genetic diagrams to predict characteristics. [H]

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Inherited conditions in humans

Learning objectives

- How are human genetic disorders inherited?
- How can we use a genetic diagram to predict whether a child will inherit a genetic disorder?
- Can you construct a genetic diagram to make predictions about the likelihood of inheriting a genetic disorder?

[H]

Not all diseases are infectious. Sometimes diseases are the result of a problem in our genes and can be passed on from parent to child. They are known as **genetic or inherited disorders**.

We can use our knowledge of dominant and recessive alleles to work out the risk of inheriting a **genetic disorder**.

- a** How is an inherited disorder different from an infectious disease?

Polydactyly

Sometimes babies are born with extra fingers or toes. This is called **polydactyly**. The most common form of polydactyly is caused by a dominant allele. It can be inherited from one parent who has the condition. People often have their extra digit removed, but some live quite happily with them.

If one of your parents has polydactyly and is heterozygous, you have a 50% chance of inheriting the disorder. That's because half of their gametes will contain the faulty allele. If they are homozygous, you will definitely have the condition.

Higher

Cystic fibrosis 

Cystic fibrosis is a genetic disorder that affects many organs of the body, particularly the lungs and the pancreas. Over 8500 people in the UK have cystic fibrosis.

Organs become clogged up by thick, sticky mucus, which stops them working properly. The reproductive system is also affected, so many people with cystic fibrosis are infertile.

Treatment for cystic fibrosis includes physiotherapy and antibiotics. These help keep the lungs clear of mucus and infections. Enzymes are used to replace the ones the pancreas cannot produce and to thin the mucus.

However, although treatments are getting better all the time, there is still no cure.

Cystic fibrosis is caused by a recessive allele so it must be inherited from both parents. Children affected by cystic fibrosis are usually born to parents who do not suffer from the disorder. They have a dominant healthy allele, which means their bodies work normally. However, they also carry the recessive cystic fibrosis allele. Because it gives them no symptoms, they have no idea it is there. They are known as **carriers**.

In the UK, one person in 25 carries the cystic fibrosis allele. Most of them will never be aware of it. They only realise when they have children with a partner who also carries the allele. Then there is a 25% (one in four) chance that any child they have will be affected.

- b** You will only inherit cystic fibrosis if you get the cystic fibrosis allele from both parents. Why?

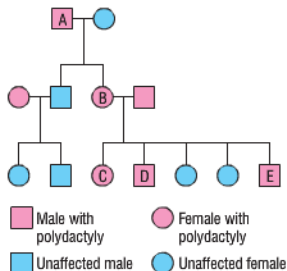


Figure 1 Polydactyly is passed through a family tree by a dominant allele



The genetic lottery

When the genes from parents are combined, it is called a genetic cross. We can show this using a genetic diagram (see Figures 2 and 3). A genetic diagram shows us:

- the alleles for a characteristic carried by the parents (the genotype of the parents)
- the possible gametes which can be formed from these
- how these could combine to form the characteristic in their offspring. The genotype of the offspring allows you to work out the possible phenotypes too.

When looking at the possibility of inheriting genetic disorders, it is important to remember that every time an egg and a sperm fuse it is down to chance which alleles combine. So if two parents who are heterozygous for the cystic fibrosis allele have four children, there is a 25% chance (one in four) that each child might have the disorder.

But in fact all four children could have cystic fibrosis, or none of them might be affected. They might all be carriers, or none of them might inherit the faulty alleles at all. It's all down to chance!

Figure 3 A genetic diagram for cystic fibrosis

	P	p
p	Pp	pp
p	Pp	pp

50% chance polydactyly, PP or Pp, 50% chance normal pp

Pp = Parent with polydactyly
pp = Normal parent

Figure 2 A genetic diagram for polydactyly

Both parents are carriers, so Cc

	C	c
C	CC	Cc
c	Cc	cc

Genotype:
25% normal (CC)
50% carriers (Cc)
25% affected by cystic fibrosis (cc)

Phenotype:
3/4, or 75% chance normal
1/4, or 25% chance cystic fibrosis

Curing genetic diseases

So far we have no way of curing genetic disorders. Scientists hope that genetic engineering could be the answer. It should be possible to cut out faulty alleles and replace them with healthy ones. They have tried this in people affected by cystic fibrosis. Unfortunately, so far they have not managed to cure anyone.

Genetic tests are available that can show people if they carry the faulty allele. This allows them to make choices such as whether or not to have a family. It is possible to screen fetuses or embryos during pregnancy for the alleles which cause inherited disorders. You can also screen embryos before they are implanted in the mother during IVF treatment. These tests are very useful but raise many ethical issues.

Summary questions

- a What is polydactyly?

b Why can one parent with the allele for polydactyly pass the condition on to their children even though the other parent is not affected?

c Look at the family tree in Figure 1. For each of the five people labelled A to E affected by polydactyly, give their possible alleles and explain your answers.
- a Why are carriers of cystic fibrosis not affected by the disorder themselves?

b Why must both of your parents be carriers of the allele for cystic fibrosis before you can inherit the disease?
- A couple have a baby who has cystic fibrosis. Neither the couple, nor their parents, have any signs of the disorder.

Draw genetic diagrams showing the possible genotypes of the grandparents and the parents to show how this could happen.

[H]

Key points

- Some disorders are inherited.
- Polydactyly is caused by a dominant allele of a gene and can be inherited from only one parent.
- Cystic fibrosis is caused by a recessive allele of a gene and so must be inherited from both parents.
- You can use genetic diagrams to predict how genetic disorders might be inherited.
- You can construct genetic diagrams to predict the inheritance of genetic disease.

[H]