Inherited Diseases
Inherited Diseases

What are inherited diseases?

Huntington's disease

Cystic fibrosis

Sickle cell anaemia

Other inherited diseases

Summary activities
What is inheritance?

Each person has a unique set of characteristics, such as eye colour, height and blood group.

A person’s characteristics are determined by a combination of the genes they inherit from their parents and the environment in which they develop.

Different versions of genes exist, resulting in variation in specific characteristics, such as different eye colours. These versions are called alleles.
**What are inherited diseases?**

**Inherited diseases** are diseases caused by faulty genetic material that is passed on to future generations. They are sometimes called genetic disorders.

Many inherited diseases are caused by mutations in DNA, resulting in faulty alleles that are not properly expressed.

Mutations can be spontaneous or caused by exposure to mutagens such as radiation and certain chemicals.

There are over 4,000 known inherited diseases, although the specific alleles involved are only known for 25% of them.
What types of inherited disease exist?

There are three main types of inherited disease:

- **Single-gene diseases**
  These are caused by mutations in single genes. Examples include Huntington’s disease, cystic fibrosis and sickle cell anaemia.

- **Multifactorial diseases**
  These are caused by mutations in several genes combined with environmental factors. Examples include heart disease, Alzheimer’s, diabetes and cancer.

- **Chromosomal diseases**
  These are caused by an abnormal number or structure of chromosomes. Examples include Down’s syndrome and Klinefelter syndrome.
What are carriers?

Does everyone who carries a faulty allele develop a disease?

Like ‘normal’ alleles, faulty alleles can be **dominant** or **recessive**.

This means that people can have a copy of a faulty allele that codes for an inherited disease, but not have the disease themselves. These people are called **carriers**.

Why is it important to know if someone is a carrier?

Carriers can potentially pass the disease on to their children.
Treating inherited diseases

Many inherited diseases are serious and even fatal, but some still enable people to lead relatively normal lives with appropriate treatment.

Most inherited diseases cannot yet be cured, however. Why is this?

Curing an inherited disease involves replacing faulty genes with normal genes.

This is called gene therapy, but it is a very complex and relatively new technique, and has yet to be widely successful.
### Inherited diseases: true or false?

<p>| | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>1.</strong></td>
<td>A person's characteristics are determined entirely by the genes that they inherit from their parents.</td>
</tr>
<tr>
<td><strong>2.</strong></td>
<td>Different versions of genes are called alleles.</td>
</tr>
<tr>
<td><strong>3.</strong></td>
<td>DNA mutations can be spontaneous or caused by exposure to radiation and certain chemicals.</td>
</tr>
<tr>
<td><strong>4.</strong></td>
<td>People with a copy of a disease-causing allele, but who don't have the disease, are called carriers.</td>
</tr>
<tr>
<td><strong>5.</strong></td>
<td>Carriers cannot pass the disease on to their children.</td>
</tr>
<tr>
<td><strong>6.</strong></td>
<td>In the future, inherited diseases may be cured by replacing faulty genes with normal genes.</td>
</tr>
</tbody>
</table>
Inherited Diseases

What are inherited diseases?

Huntington's disease

Cystic fibrosis

Sickle cell anaemia

Other inherited diseases

Summary activities
Huntington’s disease, sometimes called Huntington’s chorea, is a rare and fatal inherited disease of the central nervous system.

Huntington’s disease is caused by a single dominant allele, which means that heterozygous individuals will develop the disease.

The disease causes damage to brain cells, leading to a gradual loss of co-ordination, decline in mental ability and changes in personality.

Around 4,800 people in the UK have Huntington’s disease.
The symptoms of Huntington’s disease

Symptoms of Huntington’s disease usually begin to develop between the ages of 30 and 50, although they can appear earlier in rare occasions.

Early symptoms include:

- twitching, clumsiness and stumbling
- lack of concentration and memory loss
- depression and mood changes

Huntington’s disease is incurable, but medication can help to reduce the symptoms. However, as the disease progresses, the person’s co-ordination and movement becomes worse, and their mental abilities decline further.
How is Huntington’s disease diagnosed?

Huntington’s disease is usually diagnosed from the physical symptoms of the patient and a family history of the disease.

The diagnosis is confirmed by a blood test that detects the Huntington’s disease allele. This test can also be used in people who have yet to show symptoms but who have a family history of the disease.

Why might this be useful for someone planning to have children?

People with the Huntington’s disease allele generally develop the disease later in life, so they may unknowingly pass the allele on to their children before realizing that they have the disease.
How is Huntington's disease inherited?

Huntington's disease (HD) is caused by a **dominant** allele.

People who inherit either one or two copies of the allele develop HD.

If a male has **HD** (he is heterozygous for the disease), but a female is **unaffected**, what is the chance that their children will have HD?

Click "**start**" to find out.
What is the pattern of inheritance of HD in this family?

- Unaffected
- Carrier
- HD
Inherited Diseases

- What are inherited diseases?
- Huntington's disease
- Cystic fibrosis
- Sickle cell anaemia
- Other inherited diseases
- Summary activities
Cystic fibrosis (CF) is a common inherited disease that affects the respiratory and digestive systems by producing abnormally thick mucus.

CF is caused by a single recessive allele, which means that only homozygous individuals will develop the disease.

CF is the most common life-threatening inherited disease in the UK and other countries, affecting 1 in every 2,500 people.
The symptoms of cystic fibrosis usually develop during early childhood.

Thick mucus clogs the airways, leading to:

- breathlessness and coughs
- repeated chest infections – bacteria are trapped and thrive in the thick mucus.

The mucus also clogs the pancreatic duct and blocks enzyme secretion. This results in poor digestion, causing low weight gain and low energy levels.
How is cystic fibrosis diagnosed?

Cystic fibrosis is usually diagnosed via a sweat test.

One of the symptoms of CF is salty sweat, caused by the abnormal transport of sodium and chloride ions.

During a sweat test, a chemical and electric current are applied to a patch of skin causing it to start producing sweat. The sweat is collected and the chloride levels are tested.

A high concentration of chloride ions indicates CF.
Testing fetuses for CF

If a couple has a family history of cystic fibrosis, they may want to test their unborn child's chromosomes for the disease.

There are two types of test:

- **amniocentesis** – fetal cells are collected from the amniotic fluid via a hollow needle

- **chorionic villus sampling** – fetal cells are collected from the chorionic villi in the placenta with a suction tube.

Each procedure has a small risk of miscarriage or infection. Are the tests worthwhile? How might the results affect the parents?
How was cystic fibrosis discovered?

The discovery of cystic fibrosis

Although cystic fibrosis (CF) was not recognized as a disease until the 1930s, doctors began recording symptoms of the condition from the 1700s.

Many scientists have since contributed to increasing understanding of the disease, and how it can be effectively treated.

Click "start" to find out more.
The symptoms of CF can be effectively managed by a combination of treatments that greatly improve quality of life.

- Drugs widen the airways and break down excess mucus, while antibiotics treat chest infections.

- Oxygen gas helps improve breathing.

- Physiotherapy loosens mucus, making it easier to cough up.

- Enzyme tablets boost pancreatic enzyme levels, improving digestion and growth, and increasing energy levels.
Depending on the severity of their symptoms, most children with CF are capable of leading relatively active lives.

In fact, regular exercise is often recommended because it improves health, fitness and strength.

As people with CF get older, their lungs begin to deteriorate and some choose to go on a waiting list for a lung transplant.

The average life expectancy of a person with CF has increased as treatment improves, and is now in the mid-30s, compared with just a few months in the 1950s.
How is cystic fibrosis inherited?

Cystic fibrosis (CF) is caused by a recessive allele.

People who inherit two copies of the allele develop CF. People who inherit just one copy of the allele become carriers of the disease.

If two people are both carriers of CF, what is the chance that their children will have CF?

Click "start" to find out.
What is the pattern of inheritance of CF in this family?

- Unaffected
- Carrier
- Cystic fibrosis

solve
Inherited Diseases

- What are inherited diseases?
- Huntington's disease
- Cystic fibrosis
- Sickle cell anaemia
- Other inherited diseases
- Summary activities
Sickle cell anaemia is an inherited disease that causes red blood cells to distort and form curved, sickle shapes.

Sickle cell anaemia is caused by a single recessive allele, which produces abnormal haemoglobin – the protein that carries oxygen in red blood cells.

The disease mainly affects people of African, Caribbean, Mediterranean and Asian descent.

Over 6,000 people in the UK have sickle cell anaemia.
Common symptoms of sickle cell anaemia include anaemia, tiredness, weakness and jaundice.

These are caused the fragile nature of sickle blood cells, which only last 10–20 days as opposed to about 4 months.

In addition, people with sickle cell anaemia may have severe pain attacks called ‘crises’.

‘Crises’ are caused by sickle cells blocking blood vessels. This reduces the oxygen supply to organs, causing pain and organ damage.
There is no cure for sickle cell anaemia, but with regular medical attention most people are able to lead relatively normal lives.

During a sickle cell crisis, the person may need urgent hospital treatment.

This involves strong pain relief and may also require antibiotics, oxygen treatment and even a blood transfusion.

In the future, sickle cell anaemia may be routinely treated using bone marrow transplants or gene therapy.
What is sickle cell trait?

People who have just one sickle cell allele are carriers of the allele. They do not have sickle cell anaemia, but instead have **sickle cell trait**.

People with sickle cell trait don’t show any symptoms of the condition themselves but nevertheless have a few sickle blood cells.

In countries in Africa, Asia and Central and South America, the sickle cell allele occurs with a high frequency.

Why might this be the case?
Many countries in the developing world have a high incidence of malaria, one of the most common life-threatening diseases in the world.

Malaria is caused by infection with a parasite that lives and multiplies in red blood cells. The parasite is transmitted by mosquito bites.

What advantage might sickle cell trait offer people in countries where malaria is common?

The shape of sickle cells reduces the ability of the parasite to reproduce. This results in a resistance to malaria.
How is sickle cell anaemia inherited?

Sickle cell anaemia is caused by a **recessive** allele.

People who have two copies of the allele develop sickle cell anaemia. People who have just one copy of the allele have sickle cell trait.

If two people have **sickle cell trait**, what is the chance that their children will also inherit the condition?

Click "**start**" to find out.
What is the pattern of inheritance of SCA in this family?

- unaffected
- sickle cell trait
- sickle cell anaemia

solve
What is haemophilia?

Haemophilia is an inherited disease that prevents the body from controlling bleeding properly.

Haemophilia is a recessive sex-linked disease that mainly affects males.

People with haemophilia (haemophiliacs) are partly or completely missing a clotting factor that is needed to make their blood clot properly.

There are about 6,000 people with haemophilia in the UK.
Symptoms of haemophilia include:

- bruising easily
- prolonged bleeding
- spontaneous bleeding.

Bleeding from minor grazes and cuts can usually be stopped relatively easily using just a sticking plaster.

However, deep cuts and haemorrhaging can be very serious, affecting the joints, muscles and soft tissue.
People with haemophilia who manage their condition carefully have a normal life expectancy.

Haemophilia is treated by injections of clotting factors, although no permanent replacement is possible. Up to three injections per week may be needed.

In the past, clotting factors were taken from donated blood, but this was often contaminated with hepatitis C and HIV viruses, leading to infection of haemophiliacs.

Most clotting factor is now produced by genetically-engineering bacteria.
Haemophila in males

Haemophilia is far more common among males than females because it is a recessive X-linked disease.

Males only have one X chromosome, which is inherited from their mother.

Any allele contained within the chromosome, whether it is dominant or recessive, will always be expressed.

This is because there is only one version of the allele and its effects will therefore not be masked by another version.
Haemophilia in females

What happens if a female inherits one haemophilia allele?

\[ X^H X^h \]

= female carrier

The recessive haemophilia allele is masked by the dominant normal allele on the other X chromosome. The female is therefore not affected by the disease, but is a carrier.

What happens if a female inherits two haemophilia alleles?

\[ X^h X^h \]

= female haemophiliac

She will develop haemophilia. This is extremely rare.
How is haemophilia inherited when the mother is a carrier?

Haemophilia is caused by a recessive allele carried on the X chromosome.

If a male is unaffected, but the female is a carrier for haemophilia, what is the chance that they will have a son with haemophilia?

Click "start" to find out.
How is haemophilia inherited when the father has haemophilia?

Haemophilia is caused by a recessive allele carried on the X chromosome.

If a male has haemophilia, but a female is unaffected, what is the chance that they will have a son with haemophilia?

Click "start" to find out.
What is the pattern of haemophilia inheritance in this family?

- Unaffected
- Carrier
- Haemophiliac

solve
What is Down's syndrome?

Down's syndrome is a genetic disorder that occurs when a person inherits an extra copy of chromosome 21.

People with Down’s syndrome often have characteristic physical features, learning disabilities and have a higher risk of certain medical conditions.

The risk of having a child with Down’s syndrome increases with maternal age, especially over the age of 40.

In the UK, Down's syndrome affects about one in every 1,000 children born each year.
How is Down’s syndrome diagnosed?

During pregnancy, ultrasound and amniotic cell tests can be used to estimate the probability that a baby has Down’s syndrome.

After the baby is born, the diagnosis can be confirmed by taking a blood or tissue sample, and observing the shape and number of chromosomes under a microscope.

How would a scientist recognize Down's syndrome by looking at chromosome number?
Speech and language therapy can be very useful in helping people with Down’s syndrome communicate and express themselves.

With support, people with Down’s syndrome often go to mainstream schools, and are able to live semi-independent adult lives. Many are also able to become employed.

People with Down’s syndrome can live healthy and varied lives, and have a life expectancy of around 60 years.
Down’s syndrome and trisomy 21

What is trisomy 21?

Down's syndrome, or trisomy 21, is an inherited disease caused by the inheritance of an additional chromosome 21.

Click "start" to find out how this happens.
Inherited Diseases

What are inherited diseases?

Huntington's disease

Cystic fibrosis

Sickle cell anaemia

Other inherited diseases

Summary activities
● **allele** – One version of a gene, found at a specific location along a chromosome.

● **carrier** – An individual with a recessive allele, the effect of which is masked by a dominant allele.

● **cystic fibrosis** – A recessive inherited disease affecting the lungs and digestive system.

● **Down’s syndrome** – A genetic disorder occurring when a person inherits an extra copy of chromosome 21.

● **gene therapy** – Curing a genetic disease by replacing a faulty allele with a ‘healthy’ version.

● **haemophilia** – A sex-linked inherited disease resulting in a failure of the blood to clot properly.
● **Huntington’s disease** – A dominant inherited disease resulting in neurological problems.

● **inherited disease** – A genetic disease caused by mutations in genes or an abnormal number of chromosomes.

● **mutation** – A random change in the genetic code of a cell, often resulting in disease.

● **sickle cell anaemia** – A recessive inherited disease resulting in abnormally-shaped red blood cells.

● **sickle cell trait** – A condition in which a person is a carrier of the sickle cell gene but does not have the symptoms.
How quickly can you unscramble anagrams of words about

inherited
diseases?
Genetic causes of disease

To which genetic type do these diseases belong?

- recessive
- dominant
- sex-linked
- chromosomal

Down's syndrome
How inherent is your knowledge of inherited diseases?