Inherited Disorders
Inherited Disorders

- What are inherited disorders?
- Examples of inherited disorders
- Genetic testing
- Summary activities

GCSE Biology
What is inheritance?

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

These 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, which result in variation in specific characteristics, such as eye colour. These versions are called alleles.
Inherited disorders are caused by faulty genetic material that is passed on to future generations. They are sometimes called genetic disorders.

Many inherited disorders 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 disorders, although the specific alleles involved are only known for 25% of them.
Types of inherited disorders

There are different types of inherited disorders:

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

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

Some disorders, such as Down’s syndrome and Klinefelter syndrome, are caused when a person inherits an abnormal number or structure of chromosomes.
Fill in the gaps

What are the missing words about inherited disorders?

1. Many inherited disorders are caused by mutations in [ ] , resulting in faulty alleles that are not properly expressed.

2. There are over [ ] known inherited disorders, although the specific alleles involved are only known for 25% of them.

3. Huntington's disease is an example of a [ ] disorder.

4. [ ] and cancer are examples of
Some inherited disorders, such as Huntington’s disease, are caused by **dominant** alleles. However, most, including cystic fibrosis, are caused by **recessive** alleles.

A person can have a one faulty recessive allele that codes for an inherited disorders in a pair of alleles, but not show symptoms of the disease themselves because the normal allele is dominant. These people are called **carriers**.

If two carriers for an inherited disorder have a child, that child may go on to develop the disorder.
Treating inherited disorders

Many inherited disorders are serious and life-limiting, but some still allow people to lead relatively normal lives, with the appropriate treatment.

However, most inherited diseases cannot yet be cured. Why is this?

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

This is called **gene therapy**. It is a very complex and relatively new technique, and has yet to be widely successful.
**True or false?**

Are these statements about inherited disorders true or false?

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

What are inherited disorders?

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Symptoms of inherited disorders
Huntington’s disease (HD) is an inherited disorder caused by a dominant allele. It damages the nerve cells in the brain and eventually leads to the loss of brain function in certain areas.

HD does not develop until later in life, meaning that people may pass it on to their children before they even know they have it.
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 likelihood that their children will have CF?
Press "start" to find out.
How is sickle-cell anaemia inherited?

Sickle-cell anaemia (SCA) 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?

Press "start" to find out.
What is the pattern of inheritance of SCA in this family?
Diagnosing cystic fibrosis

Cystic fibrosis in children is usually diagnosed with a *sweat test*. However, 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.
Are these statements for or against fetal testing?

There are various implications of testing fetuses for the alleles which cause inherited disorders.

Press "start" to look at some statements for and against fetal testing.
Huntington’s disease is usually diagnosed from the physical symptoms of the patient and confirmed by a blood test.

This test can also be used in people who have yet to show symptoms but who have a family history of the disease, meaning they can start taking useful drugs before they even display symptoms.

Not everyone with a family history of an inherited disorder will want to know if they have inherited it. Why?
Couples at risk of having a baby with an inherited disorder may opt to undergo *in vitro* fertilization (IVF) with a procedure known as *pre-implantation genetic diagnosis* (PGD).

*In vitro* is Latin for ‘in glass’ so IVF means fertilization that takes place outside of the body and in laboratory glassware.

After fertilization, *embryos* can be checked for specific inherited disorders. Only embryos without the disorder will be implanted into the woman’s body.
Do you agree or disagree with these opinions on genetic testing?

Different people may have different opinions on the use of genetic testing. Discuss each statement as a class, then drag the marker to the appropriate place on the slider scale.

Press "start" to begin.

start

- disagree
- not sure
- agree
The Human Fertilisation and Embryology Authority (HFEA) is the independent body in the UK responsible for overseeing the use of embryos in fertility treatment and research.

The HFEA decides which embryo research and embryo selection should be allowed.

Science is advancing quickly, so the HFEA will increasingly have to make decisions about the ethics of new treatment and research.

If something can be done, does that mean it should be?
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Glossary of keywords: inherited disorders

allele – One version of a gene, found at a specific location along a chromosome.

amniocentesis – A prenatal test for genetic or chromosomal disorders where fetal cells are collected from the amniotic fluid via a hollow needle.

amniotic fluid – The nutritious and protective liquid that surrounds a fetus in the womb; the 'waters'.

carrier – An individual with a recessive allele, the effect of which is masked by a dominant allele.
Inherited disorders: multiple-choice quiz

Can you pass on your knowledge about inherited disorders?

Press "start" to begin.