Inherited Diseases

What is inheritance?

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

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 colors. 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 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 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**.

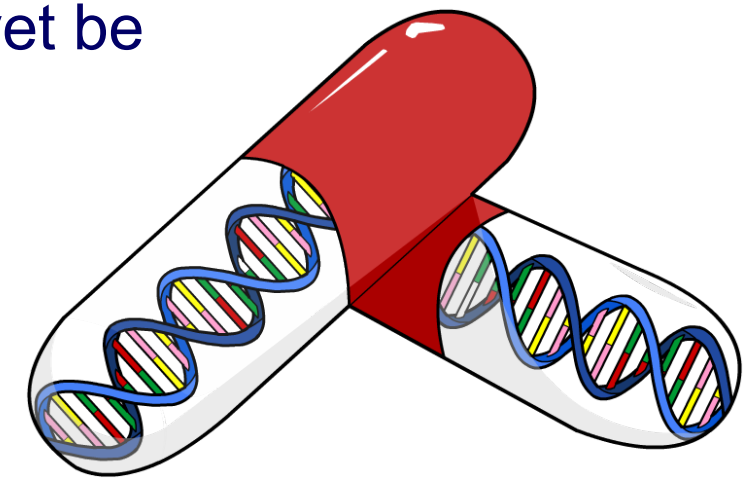
Why is it important to know if someone is a carrier?

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?