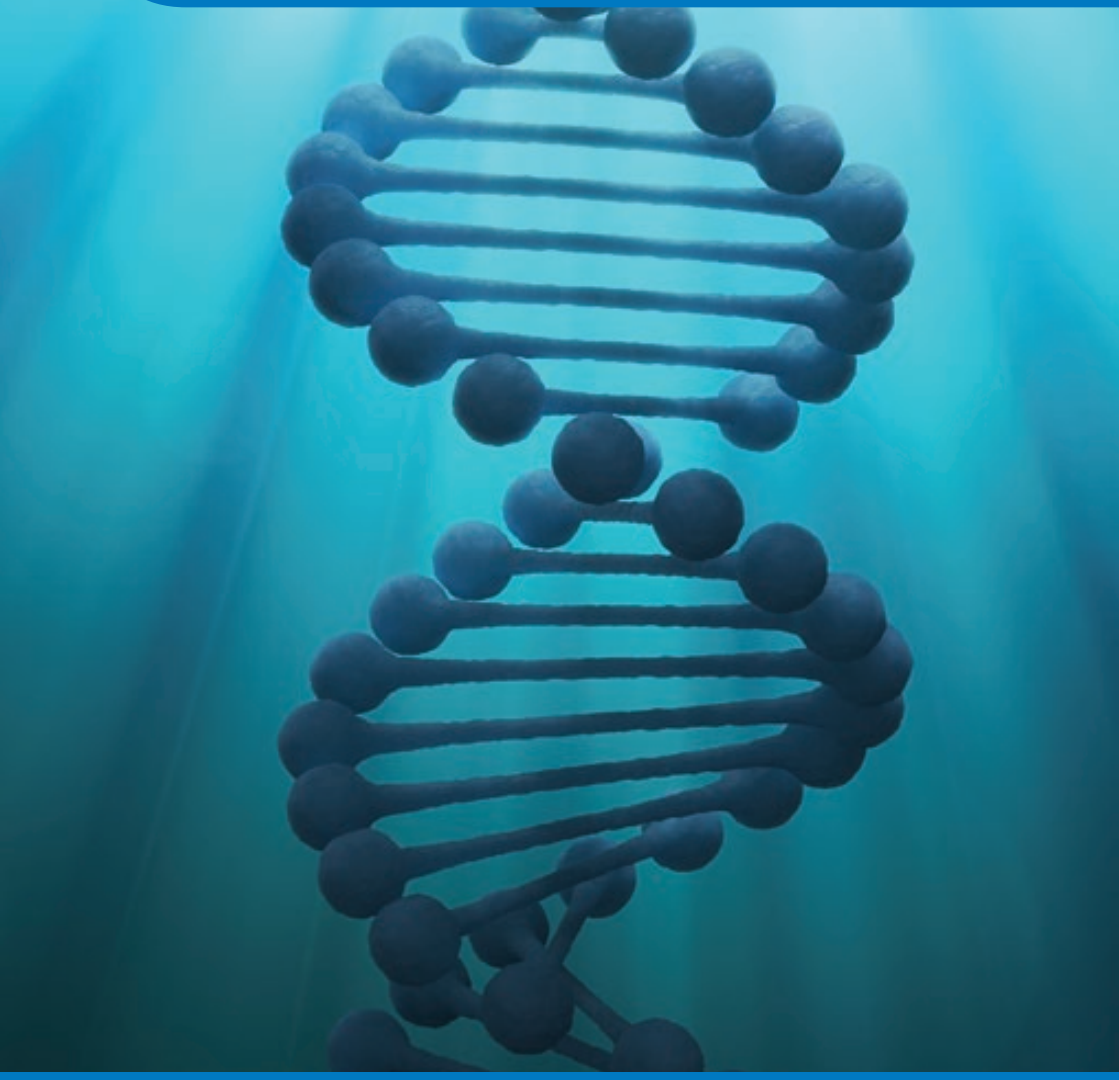
NHS
University Hospital Southampton
NHS Foundation Trust

Cystic fibrosis (CF)

Information for patients



We have written this booklet for people who have a family history of cystic fibrosis (CF) and would like to understand more about the genetics of this condition. We hope this booklet will help to answer some of the questions you may have about CF.

What is cystic fibrosis (CF)?

CF is an inherited condition that causes sticky mucus to build up in the lungs and digestive system. This causes lung infections and problems with digesting food.

CF is caused by variants in a particular gene pair. It affects around one in 2,500 children born in the UK and is usually diagnosed in childhood.

Symptoms

The build-up of sticky mucus in the lungs can cause breathing problems and increases the risk of lung infections. Over time, the lungs may stop working properly.

Mucus also clogs the pancreas (the organ that helps with digestion), which stops enzymes reaching food in the gut and helping with digestion. This means most people with CF don't absorb nutrients from food properly and need to eat more calories to avoid malnutrition.

Other symptoms of CF include:

- persistent coughing and shortness of breath
- poor growth
- diabetes
- liver problems
- osteoporosis (a health condition that weakens bones, making them fragile and more likely to break)

Treatment

Treatment for CF involves regular physiotherapy to help clear the mucus from the lungs, antibiotics to fight infection, and enzyme supplements with food to help digestion.

What are genes and chromosomes?

Genes are the unique set of instructions inside our bodies, which make each of us an individual. There are many thousands of different genes, each carrying a different instruction. We inherit two copies of each gene, one from our mother and the other from our father.

Our genes are contained on thread-like structures called chromosomes. Most people have 46 chromosomes which exist as 23 pairs. As with genes, we have two copies of each of our chromosomes, one from our mother and one from our father.

What is the CF gene?

The CF gene codes for a protein called cystic fibrosis trans-membrane conductance regulator (CFTR). This protein controls the flow of salt in and out of the cells that line the lungs and the digestive system.

Sometimes there is a variant in a gene, like a spelling mistake, that can change the instruction of the gene, preventing it from functioning correctly.

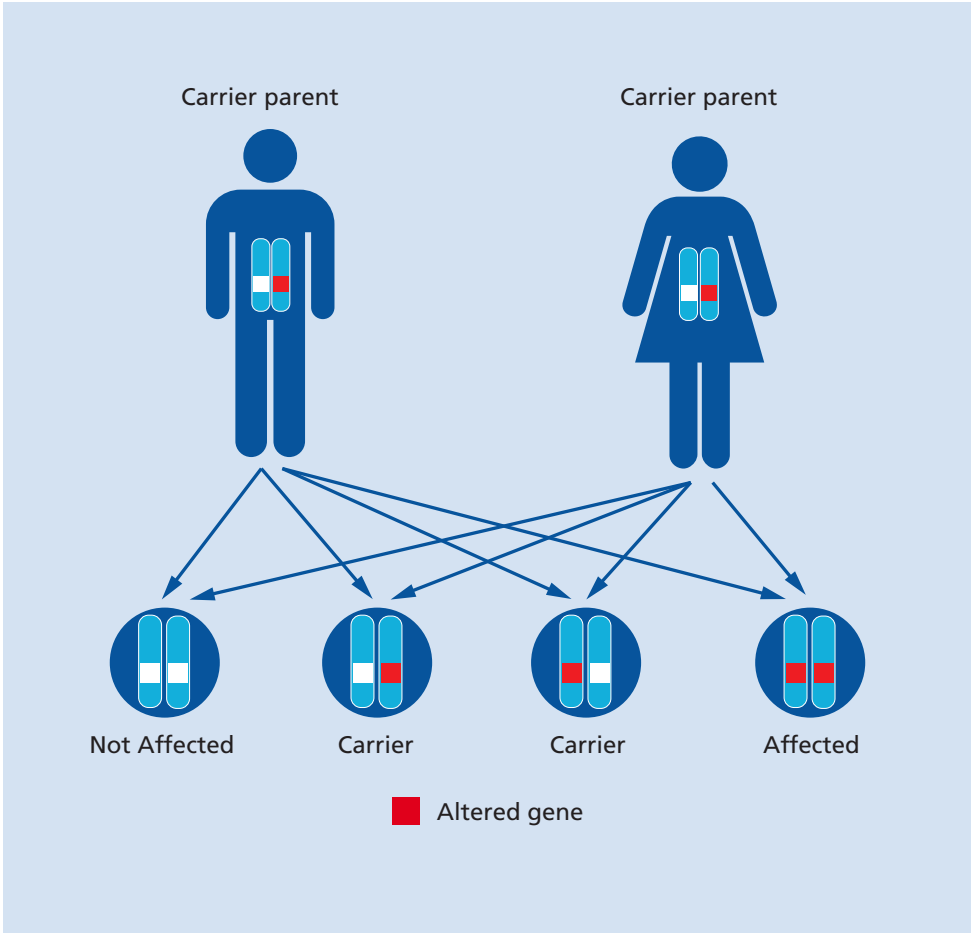
In CF, variants in the CFTR gene mean it doesn't work correctly. The mucus glands secrete too much salt and not enough water. This is the reason why the secretions of the airways are sticky and become infected, and why the sweat of a person with CF is excessively salty.

Causes

In almost every case, CF results from both parents carrying a variant in one of their two copies of the CF gene. If both healthy parents carry a variant in the CF gene, each child has a one in four (25%) chance of inheriting two gene variants, one from each parent, and therefore having CF. This is called 'autosomal recessive inheritance'. The parents are healthy gene carriers because they carry one gene copy with a variant and one normal copy. Their normal copy of the gene keeps them healthy and compensates for the copy with the gene variant.

Around one in 25 people in the white European population are carriers of CF.

Parents can sometimes misunderstand the one in four chance. If they have had one child affected by CF, they may think that a one in four chance means that the next three children cannot be affected. This is not true. The one in four chance is the same for every child the couple has (see diagram below).



Are there many variants in the CF gene?

Over 1,600 different variants have been discovered in the CF gene. However, most gene carriers in the UK have one common gene variant. This is sometimes called Delta F508, but it is more commonly known as Phe508del. The majority (94%) of people with CF carry at least one Phe508del variant.

The 53 most common variants are tested for routinely in our laboratory and they account for at least 90% of all CF variants identified in north west Europe.

If I am a carrier of a CF gene variant, can my partner have a test?

If you are found to be a carrier of one of the CF gene variants and are planning to have children, it is important for your partner to be offered carrier testing. The genetic testing currently available detects over 90% of carriers.

If we are both gene carriers, what choices do we have in pregnancy?

While there have been great improvements in the length and quality of life for people with CF, it continues to be a serious condition. Carrier parents may wish to consider their options regarding pregnancy.

With this knowledge, some people choose to continue to have children, while others choose not to have any (more) children.

Some people choose to have prenatal testing early in their pregnancy to find out if their unborn baby will have CF. There are different prenatal testing options available. These options are explained in other leaflets in this series, and a member of our genetics team would be happy to discuss them with you. It is often helpful to have considered your options before you become pregnant.

Pre-implantation genetic testing for monogenic disorders (PGT-M)

There is also the possibility of a technique called pre-implantation genetic testing for monogenic disorders (PGT-M). This is where the baby is conceived by an in vitro fertilisation (IVF) procedure and the embryos are tested for CF before they are implanted. Embryos that do not carry the CFTR gene variant and are not carriers for CF would be implanted into the woman's uterus (womb). Such tests need a lot of consideration and are not widely available.

Contact us

If you have any questions or need advice about any aspect of CF, please contact us:

- **Wessex Clinical Genetics Service**
  Princess Anne Hospital
  Coxford Road
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  SO16 5YA

  Telephone: **023 8120 6170**
  Website: **www.uhs.nhs.uk/genetics**

Further information

- **Cystic Fibrosis Trust**
  Helpline: **0300 373 1000**
  Website: **www.cysticfibrosis.org.uk**

Notes

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For help preparing for your visit, arranging an interpreter or accessing the hospital, please visit **www.uhs.nhs.uk/additionalsupport**

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