This leaflet is for people who have a family history of cystic fibrosis (CF) and would like to understand more about the genetics of this condition. It has been written to accompany a clinical genetics appointment.

**What is CF?**
Cystic fibrosis is an inherited disorder, caused by mutations in a particular gene pair. It is the most common serious genetic or inherited disease in the white European population, affecting around one in 2,500 children born in the UK. It is usually diagnosed in childhood.

**Symptoms**
People with CF produce very thick sticky mucus that affects their lungs and digestive system.

Other symptoms of cystic fibrosis include:

- persistent coughing and shortness of breath
- lung infections
- poor growth
- diabetes
- liver problems
- osteoporosis.

Treatment 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.

Genes lie on tiny structures called chromosomes. We have 23 pairs of chromosomes in each of our body cells. 22 of these pairs are the same in men and women. The 23rd pair is the X and Y chromosomes. Women have two X chromosomes, and men have one X and one Y chromosome. The cystic fibrosis gene is located on chromosome seven.

What is the cystic fibrosis gene?
The CF gene codes make 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. In cystic fibrosis the CFTR doesn’t work properly. The mucus glands secrete too much salt and not enough water. This helps to explain 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, cystic fibrosis results from both parents carrying a fault in one of their two copies of the cystic fibrosis gene. If both healthy parents carry a fault in the cystic fibrosis gene, each child has a one in four (25%) chance of inheriting two faulty genes, one from each parent and therefore having cystic fibrosis. This is called autosomal recessive inheritance. The parents are healthy gene carriers because they carry one faulty copy of a gene and one normal copy. Their normal copy of the gene keeps them healthy and compensates for the faulty copy of the gene.
Parents can sometimes misunderstand the one in four chance. If they have had one child affected by cystic fibrosis, 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 faults in the cystic fibrosis gene?
Faults in the gene are sometimes called alterations or mutations. Over 1,000 different faults have been discovered in the CF gene. However, most gene carriers in the United Kingdom (UK) have one common gene fault. This is called Delta F508. The majority (94%) of people with CF carry at least one Delta F508 mutation.

The 32 most common faults are tested for routinely in our laboratory and they account for at least 90% of all CF faults identified in northwest Europe.

If I am a carrier of a cystic fibrosis gene fault, can my partner have a test?
If you are found to be a carrier of one of the CF gene faults 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.

Some people opt to take a chance and hope the baby does not have CF. With a one in four chance, the odds are three to one in their favour. Some choose not to have any (more) children. Many choose to have prenatal testing that will tell them if the baby has inherited two cystic fibrosis genes and will therefore have cystic fibrosis.

There are choices in prenatal testing that determine early in the pregnancy whether or not a baby will have CF. These choices are explained in other leaflets in this series, and a member of the 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 diagnosis (PGD)
There is also the possibility of a technique called pre-implantation genetic diagnosis (PGD). This is where the baby is conceived by an in vitro fertilisation (IVF) procedure and the embryos tested for CF before they are implanted. Only embryos that test negative for CF would be implanted into the woman’s uterus (womb). Such tests need a lot of consideration and are not widely available.

Further information
If you need more advice about any aspect of cystic fibrosis, please contact us at:

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

  Telephone: **023 8120 6170**
  Website: [www.uhs.nhs.uk/genetics](http://www.uhs.nhs.uk/genetics)

Useful contacts

- **Cystic Fibrosis Trust**
  11 London Road
  Bromley
  Kent
  BR1 1BY

  Helpline: **0300 373 1000**
  Website: [www.cftrust.org.uk](http://www.cftrust.org.uk)