Cystic fibrosis

Cystic fibrosis (CF) is an inherited disease for which there is currently no cure. CF can have many symptoms, affecting different parts of the body, but the lungs and digestion system are most severely affected.

CF is the most common inherited genetic disease in white people, affecting about 1 in every 2,500 children born. It is much less frequent in people of African or Asian descent.

Diagnosis

About one in five babies with CF are diagnosed at birth, when their gut becomes blocked by extra thick meconium (the black tar-like bowel contents that all babies pass soon after birth). This condition may need surgery.

Just over half of people with CF are diagnosed as babies because they are not growing or putting on weight as they should. These symptoms are caused by the pancreas failing to produce chemicals (enzymes) which pass into the gut as food leaves the stomach. Without these enzymes, the fat in food cannot be properly digested. In children who are affected, the fat passes straight through the gut. The child does not benefit from the fat's energy (and so does not gain weight as he or she should). And since the stools contain an excess of fat, they are oily and very smelly.

About half of people with CF have repeated chest infections and pneumonia.

Other symptoms

CF is a "multi-system" disease, meaning that it affects many body organs. However, most of the symptoms have something to do with the lungs and the gut.

In a healthy person, there is a constant flow of mucus over the surfaces of the air passages in the lungs. This removes debris and bacteria. In someone with CF, this mucus is excessively sticky and cannot perform this role properly. In fact, the sticky mucus provides an ideal environment for bacterial growth.

People with CF are at risk of repeated bacterial chest infections. If they are not treated early and properly, these may be impossible to treat. Symptoms include persistent coughing, production of sputum (saliva and mucus), wheezing, and shortness of breath with ordinary activities.

After someone has been diagnosed with CF, if they do not have proper treatment, they will continue to have oily bowel movements, abdominal pain, and problems putting on weight. Constipation is a frequent symptom, too. Occasionally the gut becomes completely blocked, resulting in extreme stomach pain.

Other problems associated with CF can include:

- small growths (polyps) in the nose
- increased roundness of finger and toe nails with loss of the shallow groove between the bottom of the nail and skin
- an enlarged liver and spleen
- diabetes
- fertility problems in women and sterility in men

How CF is inherited

In each cell in our bodies we have 22 pairs of chromosomes and one pair of sex chromosomes. These hold the genes that determine how cells grow and function.

The abnormal gene that causes CF is found on chromosome number 7. About 1 in 20 of the white population in the UK have the CF mutation on one of the pair of number 7 chromosomes. These people are called "carriers" of the CF gene. They have no symptoms of CF - this happens only when there are CF mutations on both number 7 chromosomes (see the diagram opposite). When both parents are carriers, there is a one in four chance having either a child with CF.

There are different types of genetic mutation which are associated with different degrees of severity of the disease.

The long-term outlook

A lot of money and effort is being put into finding a cure for CF lung disease through gene therapy. However, in the meantime, children born with CF do not have normal life expectancy, though it is improving all the time. The average survival is now more than 30 years, but with the best treatment, children today with CF have a chance of better than 80% of living into their late forties.

Screening for CF

If someone has a family history of CF, they can be tested to see if they carry the CF gene before they have a family. If a couple are both carriers or if they already have a child with CF, tests can be done early in pregnancy to see if the foetus is affected. This is called chorionic villus sampling and involves taking a biopsy (a sample of tissue) from the placenta. However, there is a small risk a miscarriage with this. If the biopsy produces a positive result for CF, the parents can choose to have a termination (abortion).

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Treatment

People with CF need daily chest physiotherapy - vigorous massage to help loosen the sticky mucus. Parents of a child with CF will be taught by hospital staff how to do this. Older children and adults can be taught to do this physiotherapy on their own.

People with CF also need to have any chest infection treated quickly with antibiotics. Most people need to take capsules with meals and snacks to supply the missing pancreatic enzymes and allow proper digestion.

There is a range of other possible treatments, according to the person's condition. These may include:

- daily oral or inhaled antibiotics to counter lung infection
- inhaled anti-asthma therapy
- corticosteroid tablets
- inhalation of a medication called pulmozyme to make the sputum less sticky
- medicines to relieve constipation or to improve the activity of the enzyme supplements
- insulin for CF-related diabetes
- medication for CF-associated liver disease
- oxygen to help with breathing
- help for men with CF to overcome sterility problems

Further information

UK CF Trust
http://www.cftrust.org.uk

Cystic Fibrosis Foundation
http://www.cff.org/

Healthwise (Health Information Resource Centre)
Tel : (852) 2849 2400
Fax : (852) 2849 2900
Email : info@healthwise.org.hk
Homepage : www.healthwise.org.hk

This leaflet is for information only. For a detailed opinion or personal advice, please consult with your own doctor.

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