

- daily oral or inhaled antibiotics to counter lung infection,
- inhaled anti-asthma therapy,
- corticosteroid tablets,
- dietary vitamin supplements, especially A and D,
- 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,
- in severe cases, a lung or heart and lung transplant operation,
- help to overcome fertility problems,
- counselling to help cope with the psychological aspects.

Further information

UK CF Trust

☎ 020 8464 7211
www.cftrust.org.uk

Cystic fibrosis resource centre

www.cysticfibrosis.co.uk

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Cystic fibrosis

Cystic fibrosis (CF) is an inherited condition. It can have many symptoms, affecting different parts of the body, particularly the lungs and digestive system. CF is the most common inherited disease in white people, affecting about 1 in every 2,500 children born. It is much more rare in people of African or Asian descent.

Diagnosis of CF

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.

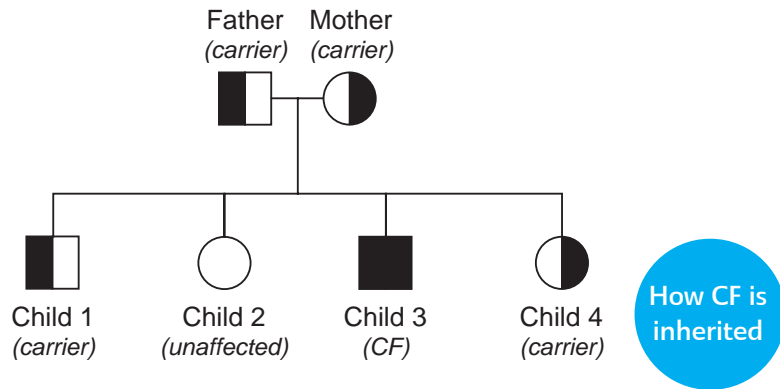
Just over half of people with CF are diagnosed as babies because they are not growing or putting on weight as they should. This is because the pancreas is not producing chemicals (enzymes) which pass into the gut as food leaves the stomach. Without these enzymes, the fat in food cannot be properly digested. In children who are affected, the fat passes straight through the gut. The child does not benefit

from the energy from the fat. Since the stools contain an excess of fat, they are oily and very smelly.

Other symptoms

CF is a "multi-system" disease, meaning that it affects many body organs. However, most of the symptoms are 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 bacterial chest infections. About half of people with CF have repeated chest infections and pneumonia. If they are not treated early and properly, these are very difficult to treat. Symptoms include persistent coughing, excess production of sputum (saliva and mucus), wheezing, and shortness of breath with ordinary activities.

If people with CF do not have proper treatment, they will continue to have oily bowel movements, abdominal pain, and problems putting on weight. Constipation is also a frequent symptom. 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 (clubbing),

- an enlarged liver and spleen,
- diabetes,
- infertility in men, because the tube that carries sperm, the vas deferens, may fail to form,
- fertility problems in women, due to thicker mucus making fertilisation difficult.

How CF is inherited

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

The abnormal gene that causes CF is found on chromosome number 7.

About 1 in 22 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 of having a child with CF, a one in two chance of having a child who is a carrier and a one in four chance of having an unaffected child.

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

The long-term outlook

Although there is currently no cure for CF, there is a lot of research under way to try to find a cure for CF lung disease through gene therapy.

Children born with CF do not have a 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 greater than 80% chance 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 fetus 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 of a miscarriage with this test. Also, if the

biopsy produces a positive result for CF, the parents then face a difficult decision of whether or not to continue with the pregnancy.

Routine newborn (neonatal) screening for CF has recently received government approval. It is done using the same blood sample that is already taken from the baby six days after birth to test for two other conditions, low thyroid function and phenylketonuria.

Treatment

People with CF need daily chest physiotherapy, which involves vigorous massage to help loosen the sticky mucus. Parents of a child with CF are taught by hospital staff how to do this. Older children and adults with CF can be taught to do this for themselves.

People with CF also need to have any chest infection treated quickly with antibiotics. The usual childhood vaccinations, such as MMR (measles, mumps and rubella) and DTP (diphtheria, tetanus and whooping cough) are important for people with CF, and they should also be vaccinated against flu and pneumococcus to help prevent chest infections.

With each meal or snack, most people with CF need to take capsules that supply the missing pancreatic enzymes and allow proper digestion.

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