Patient information from BMJ

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Cystic fibrosis: what is it?

Cystic fibrosis is a genetic (inherited) condition that is present from birth. It affects the lungs and digestive system. Treatments can help ease the symptoms and prevent complications but the condition usually gets worse as people get older. There is no cure and people with cystic fibrosis have a reduced life expectancy.

What is cystic fibrosis?

Cystic fibrosis (CF) affects the lungs and digestive system by causing them to become clogged with thick mucus so that they can't work properly.

The congestion in the lungs leads to chest infections and shortness of breath, while the mucus in the digestive organs makes it harder to digest food normally. This means that people with CF often have problems getting enough nutrition from their food.

These problems gradually get worse as people age. People with CF tend to die earlier than most people, usually because of lung problems.

CF is rare. It affects about 1 in every 3000 people. You can only get it if both your parents carry a specific faulty gene. And even then there's only a one in four chance that you will have the condition.

For information on treatments for CF, see our leaflet *Cystic fibrosis: what treatments work?*

What are the symptoms?

These days many countries screen newborn babies for CF. So if someone has CF it's usually detected at that point. The screening involves a heel-prick blood test.

If this test suggests that your baby might have CF, he or she will need further tests, including a sweat test. Testing your baby's sweat can show whether they have the condition.

The symptoms usually appear when your child is between one and three years old. But they can start earlier, when your child is still a baby, or later in childhood. Sometimes they don't really appear until adulthood.

Symptoms of CF include:

- failure to 'thrive' as a baby or young child. This means that the child doesn't seem to grow at what's considered the usual rate
- a wet-sounding cough
- shortness of breath
- being prone to chest infections
- long-term sinus problems
- an unusually large appetite.

As well as an unusually large appetite, people with CF can also have other digestive symptoms, such as diarrhoea, and very smelly stools that float because they contain a lot of fat that their digestive system can't absorb.

Clubbing' of the fingers and toes is also common in people with CF. This means that the tops of the fingernails and toenails are very rounded in shape instead of fairly flat.

If your child hasn't been diagnosed through screening at birth, and your doctor suspects CF, he or she will ask you about:

- what country your child was born in. This is to find out whether he or she was likely to have been screened at birth
- any family history of the condition
- whether your child has recurrent chest problems
- any digestive problems, such as diarrhoea, an unusually large appetite, or gastro-oesophageal reflux ('acid' reflux or heartburn).

Your doctor will also do a sweat test. If the result of this test isn't clear, and your doctor still suspects CF, he or she might suggest genetic testing.

What will happen?

CF is a serious condition that shortens people's lives. There is no cure. But the outlook is changing.

A few decades ago most people with CF died in childhood. Most people with CF now live into their late 30s, and many live much longer. And advances in treatments are still being made.

The ongoing care of people with CF is also improving. If you have CF you should be seen in a specialist centre every 3 months, or more often depending on your health. You can discuss how you are doing with a doctor and nutritionist and have tests to check if you are getting the right nutrition and how well your lungs are working.

One thing that has been shown to help people with CF stay healthier and live longer is regular exercise. Exercise helps keep the lungs working well. You can talk with your doctor about what type and how much exercise you (or your child) should do. But staying as active as possible is a good rule of thumb.

There are various charities and support groups to help people with CF and the parents of children with the condition. You should be able to find them easily online. For example, in the UK, the Cystic Fibrosis Trust (cysticfibrosis.org.uk) provides information and help with many aspects of the condition.

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