

What Is Cystic Fibrosis?

Cystic fibrosis (CF) is an inherited disease that causes the body to produce mucus that's extremely thick and sticky. The mucus is thicker than normal because CF affects cells in the epithelium (pronounced: eh-puh-thee-lee-um), the layer of cells that lines the passages in the body's organs.

In a person who does not have CF, the epithelial cells produce a thin, watery mucus that acts like a lubricant and helps protect the body's tissues. In someone with CF, however, the thicker mucus doesn't move as easily. This thick, sticky mucus clogs passages in many of the body's organs and infection sets in.

The two organs most affected are the lungs and pancreas, where the thick mucus causes breathing and digestive problems. The thicker mucus has trouble moving out of the lungs, so bacteria can remain and cause infections.

The thick mucus also is in the pancreas — an organ that produces proteins called enzymes that flow into the intestine to support the body's digestion process. Because the mucus can block the path between the pancreas and the intestines, people with CF have trouble digesting food and getting the vitamins and nutrients they need from it.

CF can also affect the liver, the sweat glands, and the reproductive organs.

What Causes CF?

Approximately 30,000 people in the United States have been diagnosed with CF, which affects both males and females. It's not contagious, so you can't catch CF from another person.

Cystic fibrosis is an inherited disease caused by mutations (changes) in a gene on chromosome 7, one of the 23 pairs of chromosomes that children inherit from their parents. CF occurs because of mutations in the gene that makes a protein called CFTR (cystic fibrosis transmembrane regulator). A person with CF produces abnormal CFTR protein — or no CFTR protein at all, which causes the body to make thick, sticky mucus instead of the thin, watery kind.

People who are born with CF have two copies of the CF gene. In almost all people born with CF, one gene is received from each parent. This means that the parents of kids with CF are usually both CF carriers — that is, they have one normal and one defective gene — but the parents may not have CF themselves because their normal gene is able to "take over" and make the necessary CFTR protein.

Each child born to parents who are both CF carriers has a 1 in 4 chance of having the disease. Cystic fibrosis occurs most frequently in Caucasians of northern European descent, in whom the CF gene is most common — although people of other heritages can get the disease, too.

People who have a close relative with CF are also more likely to carry the CF gene — approximately 12 million Americans, or 1 in every 20 people living in this country, is a CF carrier. And most of them don't know it. Parents can be tested to see if they carry the CF gene, but because there are hundreds of specific CF gene mutations (not all of which are known), genetic testing for CF won't detect everyone who is carrying a CF gene.

Doctors can also perform tests during pregnancy so prospective parents can find out more about the chances that their child will have CF. However, these tests also won't always detect a CF gene

What Happens When You Have CF?

Doctors diagnose most kids with CF by the time they are 3 years old, but if someone has a milder form of the disease, it may not be diagnosed until that person reaches the teen years. Babies are usually tested if they are born with an intestinal blockage called meconium ileus, which is more common in CF infants.

Other symptoms include:

- frequent lung infections or pneumonia
- persistent wheezing
- persistent cough with thick mucus
- bulky, light-colored, foul-smelling bowel movements or diarrhea (because food isn't being digested properly)
- failure to gain weight, even though the child eats normal amounts
- very salty sweat
- poor height growth
- nasal polyps (small growths of tissue inside the nose)
- frequent sinus infections
- fatigue

People with CF get frequent lung infections that can damage their lungs over time and require strong antibiotics along with stays in the hospital. They may have trouble growing or gaining weight because of digestive problems.

Adults with CF may also develop other illnesses, such as diabetes (when blood sugar is too high) or osteoporosis (a weakening of the bones).

The ends of the toes and fingers may become rounded and enlarged, a condition that's called clubbing.

What Do Doctors Do?

If doctors suspect that a person has CF, they will order a sweat test. In this test, the doctor or technician uses a chemical and a very mild electrical current to cause sweating on an area of skin, usually on a person's forearm (this test doesn't hurt). The doctor or technician collects a large amount of sweat in a pad and then analyzes it.

If the test results show a high level of chloride (a chemical in salt), there's a possibility the person has CF. In some states in the United States, all newborns are checked for CF with a blood test, although the blood test isn't as good at detecting the disease as the sweat test.

Doctors and people with CF can do several things to slow the progression of the disease and fight its complications. To loosen mucus, people with CF exercise regularly and may use inhalers (like kids with asthma use) or nebulizers that help deliver medication to the lungs.

Coughing helps people with CF clear the mucus from their lungs. They may also take antibiotics to prevent or fight lung infections. Chest physical therapy may also be an important part of the CF treatment routine. After lying down in a position that helps drain mucus from the lungs, the person has a helper, such as a parent, gently bang on his or her chest or back to loosen the mucus.

A newer device called a therapy vest shakes the chest, allowing teens to be more independent by doing their therapy on their own.

For digestive problems, someone with CF can take enzymes by mouth to help digest food and get nutrients from it. A doctor may also prescribe vitamin supplements and a high-calorie diet.

Living With CF

Right now, there is no cure for CF. Even if symptoms are mild at first, they get steadily worse over time.

But there is hope: 15 years ago, most children with CF would die before reaching their teens. Now, with new treatments available, more than half live into their thirties, and new research is leading to the possibility of a cure. Statistics now show that nearly 40% of the people living with CF in the United States are 18 years or older.

Since researchers identified the gene that causes CF in 1989, they have tried to replace abnormal CF genes with normal ones. Some are working on finding the right method of delivering that normal gene into the cells of a person with CF. Other scientists are trying to find new ways of fighting lung infections and different ways of moving chloride in and out of cells, bypassing the defective CFTR protein. In some cases, lung transplants have extended the lives of people with CF.

If someone you know has CF, you can be a supportive friend by learning about the disease so you can help others understand what your friend is going through. Just hanging out with your friend and taking part in the activities you enjoy doing together can be a great help.

If you have CF, focus on staying as healthy as possible by following your doctor's treatment instructions, taking your medications, eating right, and exercising. Meeting other teens who have CF via chat rooms is fine, although personal contact between two CF patients can present problems since it carries the risk of passing dangerous bacteria to each other.

Your hospital's child life and CF specialists will have all kinds of ideas and tips for coping with the disease and keeping your spirits up. Ask them to recommend groups you might join, either online or locally. They can provide you with information about clinical trials, legislative actions, handouts about CF, and even a college or vocational school scholarship program.