# WHAT IS CYSTIC FIBROSIS?



# Could You Have Cystic Fibrosis?

Most—but not all—people with cystic fibrosis have some signs and symptoms. These may include:



Wheezing



Coughing that brings up mucus or blood



Pain in the muscles or joints



Clubbing, or bulging of the tips of fingers or toes



Skin that tastes very salty

Diagnosing cystic fibrosis involves several steps. Within a few days of a baby's birth, a healthcare provider screens for the disease. If the screening result is positive, the doctor will likely perform a sweat test, which checks for high levels of chloride (a part of salt) in the sweat. If this test is positive, a doctor may do genetic testing to help select the best treatments.



#### **Basic Facts About Cystic Fibrosis**

- Cystic fibrosis affects the lung and other organs.
   The disease causes a build-up of mucus, which can lead to damage in the lungs and make it harder to breathe.
- Cystic fibrosis is an inherited disease. The parents
  of people with cystic fibrosis have both passed
  along a mutation in a gene called CFTR.
- A person can have the gene mutation and not know it. People may have the gene mutation without being aware, and without having the disease themselves. This is called being a carrier. There are about 10 million carriers in the United States.
- Anyone from any racial group can have cystic fibrosis. Many people believe that only people who are white get the disease. But cystic fibrosis sometimes gets missed in people who are Black or Hispanic, and a late diagnosis can mean worse outcomes.
- A cystic fibrosis diagnosis usually is made in early childhood. Most people with the disease get diagnosed by age 2.



**U.S. Department of Health and Human Services**National Institutes of Health





### **Managing Cystic Fibrosis**

There is no cure yet for cystic fibrosis. But people with the disease now live longer, healthier lives. Early diagnosis and treatment can make a big difference. Treatments include:

- Tips on how to cough and breathe in specific ways that help clear out mucus
- Devices that use vibrations to loosen mucus and help clear the airways
- Chest physical therapy
- Certain medications, such as:
  - Bronchodilators, or mucus thinners, as prescribed by a doctor
  - Antibiotics to reduce infections
  - New medicines that make the CFTR protein function better
- Lung transplantation, in very advanced cases

Doctors may do other tests to keep track of their patients' condition. They may also recommend the patients avoid tobacco smoke, eat a healthy diet, and wash their hands to avoid germs that can lead to infections.

## How Cystic Fibrosis Affects Your Breathing

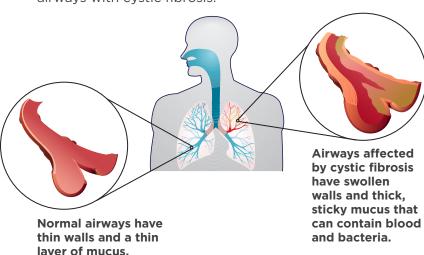
Having a mutation in the *CFTR* gene causes a protein in the body, also called CFTR, to work improperly. This affects how the body makes mucus and sweat.

With cystic fibrosis, mucus can be thick and sticky. This thick mucus can clog up the lungs and airways and make breathing difficult.

Bacteria can also grow more easily, which is why lung infections become so common.

Difficulties with clearing out mucus in the airways can lead to damage and widening of the airways. This condition is called bronchiectasis.

The image below shows normal airways, as well as airways with cystic fibrosis.



For more information about lung health, visit <a href="https://www.nhlbi.nih.gov/BreatheBetter">www.nhlbi.nih.gov/BreatheBetter</a>.

For more information about cystic fibrosis, visit www.nhlbi.nih.gov/health-topics/cystic-fibrosis.