CONDITION

# Cystic fibrosis

Learn how cystic fibrosis affects the lungs and digestion, how it is diagnosed, and how specialized treatment can protect health over time.

**WHAT IT IS**
Inherited condition causing thick sticky mucus

**BODY SYSTEM**
Lungs and digestive system

**MAIN SYMPTOMS**
Long-lasting cough, frequent lung infections, greasy stools, poor growth

**WHAT CAUSES IT**
Inherited CFTR gene changes

**HOW IT IS DIAGNOSED**
Sweat chloride and genetic testing

**MAIN TREATMENT**
Airway clearance, infection control, nutrition support

**CAN IT BE CURED?**
No, treatments do not cure it

**IS IT AN EMERGENCY?**
Sometimes, with severe difficulty breathing

**LONG-TERM OUTLOOK**
Variable with possible progressive lung decline

## Overview

Cystic fibrosis, usually called CF, is an inherited condition that affects how salt and water move in and out of cells. It is caused by changes in the CFTR gene . These changes can make mucus unusually thick and sticky.

Thick mucus can build up in the airways, making it harder to clear germs and debris from the lungs. This can lead to repeated infections, inflammation, and gradual lung damage. CF can also block ducts in the pancreas, an organ that releases enzymes needed to digest food. As a result, some people have trouble absorbing fat, protein, vitamins, and calories.

CF affects each person differently. Some people develop symptoms in infancy, while others are diagnosed later and may have milder or less typical disease. Treatments now address several parts of CF, including mucus clearance, infection control, nutrition, and the faulty CFTR

protein itself. Regular care through a specialist CF center can help protect lung function and manage complications.

## Symptoms

CF symptoms vary with age, the organs involved, and the specific CFTR gene changes. Newborn screening may identify CF before symptoms appear.

Possible breathing and sinus symptoms include:

Digestive symptoms often develop when mucus prevents pancreatic enzymes from reaching the intestines. These symptoms may include frequent, bulky, greasy, or strong-smelling stools; abdominal swelling; constipation; poor growth; or difficulty gaining weight despite a good appetite.

Some newborns with CF develop meconium ileus , a blockage caused by unusually thick first stool. This may cause abdominal swelling, vomiting, or failure to pass stool and needs prompt medical care.

People with CF lose more salt in their sweat. During hot weather, exercise, fever, or illness, this may contribute to dehydration or a low blood salt level. Symptoms can include headache, weakness, muscle cramps, nausea, or unusual tiredness.

Other possible signs include clubbing, in which the ends of the fingers become wider and rounder, and reduced fertility. Most males with CF have blocked or missing tubes that normally carry sperm. Some females may have thicker cervical mucus, but pregnancy is still possible.

These symptoms can occur in many conditions. Their presence does not by itself mean that someone has CF.

- A long-lasting cough, sometimes with thick mucus
- Wheezing or shortness of breath
- Frequent or difficult-to-clear lung infections
- Ongoing nasal congestion or sinus infections
- Nasal polyps, which are soft growths inside the nose
- Reduced ability to exercise compared with usual

## Causes and risk factors

CF is caused by disease-causing variants, or changes, in both copies of the CFTR gene. This

gene provides instructions for a protein that works as a channel for chloride, a part of salt. When the channel does not work properly, the balance of salt and water at cell surfaces changes. Mucus then becomes thicker and harder to move.

CF follows an autosomal recessive inheritance pattern. This means a person generally must inherit one disease-causing CFTR variant from each biological parent to have CF. A person with only one such variant is usually called a carrier and generally does not have cystic fibrosis.

When both biological parents are carriers, each pregnancy has:

These chances start again with every pregnancy. They do not change based on the outcomes of earlier pregnancies.

CF can occur in people of any racial or ethnic background. It is more common in some populations, but relying on ancestry or appearance can delay diagnosis. A family history of CF or known CFTR carrier status increases the chance that a person may have or carry a CFTR variant.

CF is not contagious. It cannot be acquired from another person, diet, parenting, or environmental exposure.

- A 1 in 4 chance that the child will have CF
- A 1 in 2 chance that the child will be a carrier
- A 1 in 4 chance that the child will inherit neither familial variant

## Diagnosis

In the United States, newborns are routinely screened for CF. Screening usually begins with a blood test that checks for a substance associated with pancreatic stress. Some programs also test for common CFTR variants. A positive newborn screen means further testing is needed; it is not a diagnosis by itself.

The sweat chloride test is the main diagnostic test. A trained team stimulates a small area of skin to produce sweat, collects the sweat, and measures its chloride level. The test is not painful, although the skin may briefly tingle or feel warm. Results are interpreted using established ranges, the person's age, symptoms, and other test findings. An intermediate or unclear result may require repeat testing.

Genetic testing can look for CFTR variants. A standard panel may not detect every disease-causing change, so broader genetic analysis may be considered when CF remains possible despite limited initial findings.

If sweat and genetic results do not give a clear answer, a specialist center may perform tests

that directly measure CFTR function in cells lining the nose or intestine. These tests are available only at certain centers.

After diagnosis, the care team evaluates how CF is affecting the body. Testing may include breathing tests, respiratory cultures, chest imaging, blood tests, nutrition assessment, stool testing for pancreatic function, and screening for CF-related diabetes or liver disease.

Some people have a CFTR-related disorder , meaning CFTR dysfunction affects one or a few organs without meeting full diagnostic criteria for CF. Specialist assessment is important because diagnosis affects monitoring, treatment, and family counseling.

## Treatment and management

CF treatment is usually coordinated through an accredited specialist center. The care team may include a pulmonologist, nurse, respiratory therapist, dietitian, pharmacist, social worker, physical therapist, and other specialists.

Airway-clearance therapy helps loosen mucus so it can be coughed out. Methods include chest physiotherapy, breathing techniques, positive-pressure devices, and inflatable vests. The best method and schedule depend on age, health, and personal circumstances.

Inhaled medicines may be used before or during airway clearance. Depending on the person's needs, these may include medicines that open the airways, concentrated saline that draws water into mucus, or dornase alfa, which makes mucus less thick. A CF team can explain the correct order, cleaning process, and infection-control steps for nebulizers and other equipment.

Antibiotics like amoxicillin or doxycycline treat or suppress bacterial infections. They may be inhaled, taken by mouth, or given through a vein. The choice depends on respiratory culture results, symptoms, prior response, allergies, and illness severity. Long-term use of certain antibiotics may be appropriate for selected patients under specialist supervision.

CFTR modulators improve the function of the faulty CFTR protein. Different modulators work for specific CFTR variants, and eligibility can also depend on age and local regulatory approval. These medicines do not cure CF, and they do not replace airway, nutrition, or infection care. They may require laboratory monitoring and review for medicine interactions.

Many people with CF have pancreatic insufficiency, meaning the pancreas does not release enough digestive enzymes into the intestine. Pancreatic enzyme replacement capsules are taken with food to improve digestion. The care team adjusts treatment based on growth, weight, symptoms, and stool pattern rather than using one plan for everyone.

Nutrition care may include energy- and protein-rich foods, extra salt in selected situations, and

supplements of vitamins A, D, E, and K when prescribed. Individual needs can change after starting highly effective CFTR modulator treatment, so ongoing dietitian review remains important.

Routine vaccinations help reduce preventable infections. Regular physical activity can support fitness, bone health, mucus clearance, and emotional well-being. CF-related diabetes, liver disease, bone disease, fertility concerns, anxiety, and depression each need condition-specific care.

People with advanced lung disease may be evaluated for lung transplantation. Referral is an opportunity to learn about options and timing; it does not mean transplantation will happen immediately.

## Complications

Repeated infection and inflammation can damage and widen the airways, a condition called bronchiectasis . Over time, lung function may decline. Some people cough up blood, develop a collapsed lung, or need supplemental oxygen or breathing support.

A pulmonary exacerbation is a worsening of respiratory health. Possible signs include increased cough or mucus, a change in mucus color, fever, reduced appetite, weight loss, lower exercise tolerance, or a drop in lung function. Treatment may require more airway clearance and antibiotics, sometimes in a hospital.

Digestive complications can include poor absorption of nutrients, slowed growth, constipation, intestinal blockage, pancreatitis, gallstones, and liver or bile-duct disease. CF-related diabetes has features of both type 1 and type 2 diabetes and requires specific screening and treatment.

Low bone density and fractures may result from poor vitamin absorption, inflammation, reduced activity, or other factors. Sinus disease, chronic pain, sleep problems, urinary leakage with coughing, and fertility difficulties can also affect quality of life.

CF care can create a substantial emotional and practical burden. Screening for anxiety and depression is part of comprehensive care because mental health can affect daily treatment, relationships, school, work, and physical health.

## Follow up

Routine follow-up usually includes regular visits with the CF care team, even when the person feels well. Visits may involve weight and growth measurements, breathing tests, respiratory cultures, treatment review, and discussion of nutrition, exercise, sleep, mental health, school,

work, or family planning.

Periodic blood tests and other assessments may monitor vitamin levels, liver health, blood sugar, bone health, medication effects, and complications. The exact schedule depends on age, symptoms, treatments, and clinical findings.

Contact the CF team promptly for a clear increase in cough or mucus, fever, reduced appetite, weight loss, unusual fatigue, new shortness of breath, vomiting, worsening constipation, or reduced ability to complete usual activities. Early review may prevent a manageable problem from becoming more serious.

Seek urgent or emergency care for severe difficulty breathing, blue or gray lips, fainting, confusion, coughing up more than a small streak of blood, severe dehydration, sudden chest pain with breathlessness, persistent vomiting, or severe abdominal swelling with inability to pass stool or gas. Follow the emergency plan provided by the person's CF team.

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