

CYSTIC FIBROSIS (CF)

Cystic fibrosis (CF) is an inherited disease that causes thickened mucus to form in the lungs, pancreas and other organs. In the lungs, this mucus blocks the airways, causing lung damage and making it hard to breathe. CF is a life-threatening condition, but thanks to advances in treatment and care, the average life expectancy has been steadily increasing and quality of life has improved.

Learn About Cystic Fibrosis

Cystic fibrosis is a genetic (inherited) condition that leads to recurrent sinus and pulmonary infections, as well as gastrointestinal problems.

Key Facts

- Cystic fibrosis is genetic.
- People with this condition either don't make enough or make an abnormal version of a protein called cystic fibrosis transmembrane regulator (CFTR).
- CFTR is present on the cell surface in many organs and regulates the movement of salt-sodium (Na) and chloride (Cl) ions, as well as water across the cell surface.

What Is Cystic Fibrosis?

Cystic fibrosis (CF) is a genetic disease, meaning it is passed on from parents to their children through their genes. Both parents must pass on the abnormal gene (be a carrier, ie, they each have one abnormal gene) for a child to be at-risk of developing the disease.

Cystic fibrosis affects a specific protein called cystic fibrosis transmembrane regulator (CFTR) that controls the normal movement of sodium (Na), chloride (Cl), as well as water in and out of the cells in different parts of the body. People with CF either have too little or abnormal CFTR.

When CFTR is absent or defective, the mucus normally secreted by the cells in the pulmonary airways (breathing tubes), pancreatic ducts, gastrointestinal tract, and reproductive system becomes thickened. This thickened mucus causes blockages (obstructions), frequent infection, and loss of function in the affected organs.

How Cystic Fibrosis Affects Your Body

Cystic fibrosis can affect the whole body.

- In the lungs, malfunction of CFTR leads to thick mucus that obstructs the airways. When mucus is not cleared, it creates an environment for bacteria to grow and infect the airways. This leads to a vicious cycle with more mucus, more airway blockage, and more infections. Over the course of many years, this damages the airways and eventually the lung tissue.

CF also affects other organs and systems in the body. For example:

- In the pancreas, ducts become blocked, causing fibrosis (scarring) of the pancreas. This leads to reduced absorption of fat and vitamins. And it can also cause a type of diabetes referred to as "cystic fibrosis-related diabetes."
- In the digestive tract, intestinal secretions can be much thicker than normal. This causes blockages in the intestines and sometimes requires surgery.
- In the liver, ducts can be blocked causing damage to the liver cells and cirrhosis.
- In the reproductive tract, increased mucus can cause decreased fertility or infertility in both men and women.

How Serious Is Cystic Fibrosis?

CF is a life-threatening condition. There are about 30,000 people with cystic fibrosis in the United States and approximately 70,000 people worldwide. Approximately 1 in 30 Americans is a carrier. Sixty years ago, children diagnosed with CF usually did not survive childhood. However, due to improved care, the average life expectancy has been steadily increasing since the 1950s and is currently close to 40 years. Almost half of those affected with CF are now over 18 years of age. Although it is seen in all racial groups, it is most common in Caucasians, and rare in individuals from the Far East and Native Americans.

Cystic Fibrosis Symptoms, Causes, and Risk Factors

Previously, most people with CF were diagnosed by the age of 2 because of symptoms. In the last decade, newborn screening has become available and is now available in all 50 US states. This means that infants are diagnosed before they have symptoms so that they can begin receiving care as early as possible.

What Are the Symptoms of Cystic Fibrosis?

You or your loved one with CF might experience a wide range in severity of CF symptoms. Even within the same family, siblings can have differing disease severity.

Symptoms of CF can be classified into two main categories: symptoms of respiratory tract disease and symptoms of gastrointestinal disease.

The most common symptoms of CF respiratory tract disease are:

- Chronic coughing (dry or coughing up mucus)
- Recurring chest colds
- Wheezing (that may not respond to standard asthma therapy)
- Shortness of breath
- Frequent sinus infections
- Allergies that last all year

Symptoms of lung disease can start in infancy, especially following upper respiratory viral infections. You'll experience a small but progressive loss in lung function with every passing year, leading to increased symptoms as you age.

The most common symptoms of CF gastrointestinal disease are:

- Frequent large, greasy, and foul-smelling bowel movements
- Inability to gain weight despite being hungry all of the time
- Poor growth
- Constipation and intestinal blockage
- Recurrent inflammation of the pancreas (pancreatitis)
- Symptoms of high blood sugar, such as being thirsty and urinating frequently

Other symptoms:

- Excessive sweating
- Your child with CF may taste "salty" when kissed.

What Causes Cystic Fibrosis?

CF is an inherited disorder caused by having two abnormal copies of the CF gene. You cannot catch or acquire CF. It can occur in people who have no known family history of the disease, because people with one abnormal CF gene (called "carriers") are usually healthy.

What Are the Risk Factors?

The only risk factor for getting CF is having two parents who carry abnormal CF genes and pass the abnormal gene to their child. However, there are factors that impact how severe the CF is.

- **Genes:** CF gene mutations are divided into classes based on how damaged the CFTR protein function is. Classes I, II, and III are generally more severe causing “classic CF.” Classes IV and V are usually milder. Also, other genes called modifier genes can affect a person’s symptoms and outcome.
- **Environment and lifestyle:** People with CF need to consume a very large number of calories to maintain weight and grow, which can be difficult to achieve. Physical activity is also important to help keep lungs healthy. People with CF should not smoke or be exposed to secondhand smoke, as it will worsen lung disease. You should also be careful with alcohol intake, and avoid it altogether if you have liver disease.
- **Age:** CF worsens with age. If you have CF, you usually experience a small decline in lung function each year.

When to See Your Doctor

If you or a loved one have a family history of CF and have pulmonary symptoms, have been diagnosed with CF, or experience these symptoms, consult your health-care provider, and request an evaluation at an accredited CF center. The Cystic Fibrosis Foundation website has a tool to assist in locating a care center.

Diagnosing and Treating Cystic Fibrosis

In the last 2 decades, there have been many advances in the treatment of CF. There are medications to help thin and clear the thick mucus from the airways, enzymes to help absorb fat and nutrients, and antibiotics to treat the infections. There are also new emerging treatments that target fixing the CFTR protein.

What to Expect

The new classes of CFTR-modifying medications not only improve lung function in older patients but also maintain it when given to younger patients. People with CF will still need to continue taking enzymes to help absorb food and will periodically need treatment with antibiotics for infections, but the frequency should be much less.

How Cystic Fibrosis Is Diagnosed

- **Newborn screening.** The newborn screen shows infants who have a high level of an enzyme called immunoreactive trypsin in their blood. This occurs when there is injury to the pancreas. The test is repeated if it is abnormal. Some states also combine this with testing for the most common gene mutation called deltaF508. The next step is to refer the infant for further testing as there are many “false-positive” tests. This entails taking a blood sample to check whether the infant has two genes that cause CF and/or performing a sweat test.
- **Genetic testing.** More than 2,000 gene mutations have been described in CF. Most of them are quite rare, but a few are common, like the deltaF508 mutation that is found in at least one gene in 70% of individuals. In most cases, genetic testing is able to determine the exact mutation in most cases. For couples who want to have children, genetic testing is also important as more than 10 million Americans are carriers of a CF gene. For every pregnancy, there is a one-in-four chance that the child will have CF when both parents are carriers.

- **Sweat test.** Sweat is collected from a small area on the child's forearm, and the sodium levels are measured. Children with CF have high levels of sodium in their sweat because a lack of CFTR prevents the salt on the skin from being reabsorbed back into the sweat glands.
- **Measuring nasal lining.** Another way to confirm the diagnosis is to run a small electrical current across the nasal lining (epithelium). Different solutions are applied to the nasal lining and the electrical current is measured. People with CF respond very differently than those without CF to this test, and it may help confirm a diagnosis.

How Cystic Fibrosis Is Treated

Treating the respiratory tract is very important to prevent or slow down the long-term lung damage from CF.

Airway Clearance Therapy

People with CF need to perform "airway clearance therapy" (ACT). This can be done using manual chest physiotherapy or a device called the "VEST." This jacket vibrates or "flutters" when you breathe through a device. This shakes the mucus in the airways, enabling you to cough it up.

Mucus Thinning Medication

A nebulizer or inhaler is often used before performing ACT. Commonly used medications are albuterol that relaxes the airway and helps mucus to clear, dornase alpha that thins the mucus so that you can cough it up more easily, and hypertonic saline solution that restores moisture and salt to the cell surface in the airway.

Enzymes and Nutrients

Other medications that are very important are pancreatic enzyme replacement therapies. These help the body absorb food and necessary nutrients. Enzymes have to be given before every meal or snack. People with CF also have to take certain vitamins to help absorb fat. Additional salt also needs to be provided in formula or in food.

Antibiotics and Antiinflammatories

Antibiotics are frequently needed to treat bacteria that grow in the mucus. These can be given in one of three ways:

- Orally (by mouth) – this is the easiest and cheapest route.
- By inhalation – this is more expensive but very effective.
- Intravenously (IV) – this is usually reserved for those who are sicker.

Antiinflammatory medications have also been found to be helpful in CF. Two medications are currently in use, ibuprofen and azithromycin (an antibiotic that's used as an antiinflammatory agent in CF).

In 2015, a combination medication called ivacaftor/lumacaftor was approved for children ages 12 years and older. This medication helps individuals who carry two genes of the commonest mutation deltaF508 (Class II).

More medications are in clinical trials and expected to be approved in the near future. The medications currently being tested will help those who carry the most common mutation deltaF508 (Class II) and individuals who do not make CFTR protein at all (Class I).

Living With Cystic Fibrosis

A diagnosis of CF is life changing for a family. However, in the last 2 to 3 decades, significant strides have been made so that children born today with CF have a completely different outlook than those born 30 or 40 years ago. People with CF are not only living much longer but also have a better quality of life.

What to Expect

You will meet with the care team (physicians, CF care coordinator, nurses, respiratory therapists, nutritionist, and social worker) who will explain the condition, map out a CF care strategy, and discuss resources.

Some children will need baseline lab work or x-rays at the first visit, depending on their age. Throat cultures are obtained four to six times per year. Newborns are seen very frequently until a feeding regimen, enzyme dosage, and weight gain are established.

Hospitalizations may be required for complications, such as intestinal blockage or pulmonary exacerbations. Ongoing medical treatment tries to keep these to a minimum, and address them in the outpatient setting whenever possible.

Managing Cystic Fibrosis

There are CF centers on nutrition and lung health to help manage the disease.

Maintaining normal weight is very important, as it has been shown to be linked to lung health. A person with CF needs to take in adequate calories. This is sometimes hard to achieve without the use of nutritional supplements or other means, such as a feeding tube. You may also need to take in more salt, especially if you are sweating. An appropriate enzyme and vitamin dose is also very important in the management of CF.

People with CF usually need to do daily airway clearance therapy (ACT) and may need to use inhaled medications or even antibiotics when necessary. It is also important to exercise and stay active for your lung health. You should also make sure any respiratory tract infections are treated promptly. If you think you have an infection, call your doctor, so you can receive medication.

Finding Support

The Lung Association recommends patients and caregivers join our Living with Lung Disease Support Community to connect with others facing this disease. You can also call the Lung Association's Lung Helpline at 1-800-LUNGUSA to talk to a trained respiratory professional who can help answer your questions and connect you with support.

Parent support groups and the local CF chapters of the CF Foundation are also available to help support parents of newly diagnosed CF in children, as well as in older individuals with CF. In addition, the social worker on the CF care team provides counseling on many issues, including:

- School concerns - schools need to be aware of the child's diagnosis so that appropriate measures are put in place and the child can receive the support they need.
- Insurance – advice on policy selection and additional resources to make the medications used to treat CF affordable.
- Jobs - advice on communicating with your employer, retirement, and disability.

The CF Patient Assistance Foundation helps people with CF afford medication and the devices needed to stay healthy. It also assists individuals in applying for supplemental social security and social security disability benefits.

There is also a CF legal hotline that provides information for individuals with CF about their rights.

Questions to Ask Your Doctor About Cystic Fibrosis

Making notes before your visit, as well as taking along a trusted family member or friend, can help you through the first appointment with your doctor.

- My newborn infant has been diagnosed with CF. Can I still breast-feed him?
- Can my infant with CF go to daycare?
- How can my child with CF receive enough nutrition and his or her enzymes during the school day?
- Can my child with CF be in the same classroom as another child with CF?
- Can we have pets at home if my child has CF?
- I have two children with CF. Do I need to keep them apart?
- How long do people with CF live?
- Can a lung transplant cure CF?
- Do people with CF need their treatment daily or can they skip days?

Author

Iman Sami, MD

chestfoundation.org/patienteducation