



## Genetic Fact Sheets for Parents

# Other Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

**Disorder name: Cystic Fibrosis**  
**Acronym: CF**

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This fact sheet has general information about cystic fibrosis (CF). Every child is different and some of this information may not apply to your child specifically. Certain treatments may be recommended for some children but not others. If you have specific questions about CF and available treatments, you should contact your doctor.

### What is CF?

Cystic fibrosis (CF) is an inherited condition that causes problems with lung function, and also, often, with digestion. CF causes thick, sticky mucus and fluids to build up in certain organs in the body, especially the lungs and the pancreas. When glands and organs in the body become blocked, their normal functions slow down or stop working well. This results in chronic health problems.

In people with CF, the thickened mucus that lines the lungs and bronchioles can lead to repeated lung infections. In people who do not have CF, thin slippery mucus normally lines the nose and the tubes leading to the lungs. This mucus has the job of picking up bacteria, viruses, and dirt from the air we breathe and moving them up and out of the lungs. The thick, sticky mucus found in people with CF can no longer do this job well. CF also reduces the immune cells' ability to fight infections. People with CF develop chronic coughing and recurrent lung infections.

In addition to lung problems, many children with CF also have 'pancreatic insufficiency.' The pancreas is an organ behind the stomach. One of its jobs is to make special digestive enzymes that break down the food we eat into nutrients small enough to get into the blood. If the pancreas is blocked, the enzymes cannot get to the small intestine to do their job. Without digestive enzymes, food in the small intestine cannot be broken down properly and nutrients cannot be absorbed. This often leads to poor growth and poor weight gain. It can also cause sluggishness and anemia. Because fat is not absorbed well, it ends up in the stools and causes them to be bulky, lighter in color, and have a stronger odor.

## **What causes CF?**

CF is an inherited condition that occurs when a particular cell protein is either missing or not working well. This protein is called "cystic fibrosis transmembrane conductance regulator" (CFTR). CFTR is normally made by the body and is not something we get by eating. One of CFTR's jobs is to let chloride (a molecule found in salt) in and out of the cells of the body. Researchers are still trying to find out more about why the lack of CFTR causes the health problems seen in people with CF. People with CF have a CFTR protein that is either missing or not working well.

CF is not contagious. You cannot get CF from living with, touching, or spending time with a person with CF.

## **What are the symptoms of CF?**

CF is variable and causes minimal effects in some people and more serious health problems in others. Symptoms usually start in early childhood. In fact, most children with CF show effects before one year of age. There are some people who do not find out they have CF until adulthood.

The first things parents often notice when a child has CF are:

- Salty sweat; many parents notice a salty taste when kissing their child
- Poor weight gain and growth, even when a baby or child eats a lot. This is sometimes called "failure to thrive" (FTT)
- Constant coughing or wheezing

- Thick mucus and phlegm
- Many lung and sinus infections (pneumonias and bronchitis)
- Greasy, smelly stools that are bulky and pale colored
- Intestinal problems (diarrhea or constipation, pain, gas)
- Polyyps in the nose

About 15-20% of newborns with CF have a blockage of their intestines called meconium ileus. This is caused by thick stool that gets stuck in the intestines.

About 15% of children with CF have lung effects but do not have problems with digestion. About 85% of children have problems with both lungs and digestion. There are also some people who have been diagnosed with CF because of genetic test results, but who have very few symptoms of CF.

Over time, people with CF can have chronic health issues such as:

- Repeated bouts of bronchitis or pneumonias leading to permanent lung damage
- Collapsed lung, bleeding from the lungs, or lung failure
- Poor growth and poor weight gain due to malnutrition
- Chronic diarrhea
- Fatigue and anemia
- Males are usually infertile due to blocked or absent vas deferens (the tubes carrying the sperm from the testes to the penis). There are now assisted reproductive techniques which allow some men with CF to father their own children.
- A small number of people with CF develop high blood sugar and may need insulin therapy
- Some people with CF have bouts of pancreatitis, a painful inflammation of the pancreas
- Some people with CF develop liver disease over time
- Bone thinning, also known as osteoporosis, is seen in some people with CF

If treated appropriately, CF does **not** affect intelligence or the ability to learn. People with CF can attend regular school and should be able to achieve the same level of education as people who do not have CF. Many people with CF have finished college and have full-time jobs.

If left untreated, CF can cause serious chronic health effects that could lead to early death. Many of the symptoms of CF can be controlled with proper medication and treatment. Lung infection or permanent damage to the lungs is the main cause of death in people with CF.

It is important that you see your doctor and follow a treatment plan tailored for your child's needs.

## What is the treatment for CF?

Children and adults with CF are usually treated by a team of doctors and other health care providers who have experience with cystic fibrosis. These teams are often located in special CF treatment centers. There are many CF treatment centers located throughout the US. You can find a center in your area through the Cystic Fibrosis Foundation ([www.cff.org](http://www.cff.org)).

The main goal of treatment is to keep your child's lungs clear of thick mucus and to provide your child with the correct amounts of calories and nutrients to keep him or her healthy.

Certain treatments may be advised for some children but not others. When necessary, treatment is usually needed throughout life. The following are treatments sometimes suggested for children with CF:

### 1. Pancreatic enzymes

People with CF who have blockage of the pancreas (also called "pancreatic insufficiency") need to take digestive enzymes in capsule form. These enzyme capsules need to be taken before each meal or snack. The enzymes will help your child digest food properly and allow him or her to gain weight and grow at a healthy rate.

Babies with CF can sometimes have 'failure to thrive,' a condition in which their weight and height is far below that is expected for their age. Pancreatic enzymes, along with a carefully planned diet, will help treat failure to thrive and will help your baby to grow at a healthier rate.

### 2. Diet and Vitamins

- **Vitamin supplements:** People with CF have trouble absorbing some vitamins, especially fat-soluble vitamins such as vitamin A, D, E, and K. Specific supplements may be suggested for your child.
- **A higher-calorie diet:** Many babies and children with CF need more food than typical in order to stay healthy. Some children with CF need up to twice the normal number of calories to grow appropriately. A dietician who has experience with CF can help you come up with a good nutrition plan for your child.
- **Extra fluids:** Your child may need to drink more water and liquids than other children in order to help loosen the thick mucus and to prevent dehydration. Children with CF lose more salt than others, especially during exercise or in hot weather.

### **3. Airway clearance therapy**

Airway clearance therapy is done to break up and move mucus that has settled in the lungs and bronchi so that it can more easily be coughed up. It is usually performed several times a day and takes up to 20 to 30 minutes for each session. There are a number of ways to perform airway clearance therapy. Your doctor will recommend a method that will be most effective for you and your child. Some common types of airway clearance therapy are:

- **Chest percussive therapy:** Some people with CF have a parent or caregiver tap or clap on their chest and back to break up and move mucus. Some people use a handheld machine that causes vibrations on the chest and back.
- **ThAirapy vest:** Some people use a special vest that vibrates to break up the mucus.

### **4. Medications**

Your doctor may recommend special medications to treat the lung symptoms of CF. Examples of these medications include:

- **Bronchodilators:** These are inhaled drugs that open the airways to the lungs
- **Mucus thinners:** These are inhaled drugs that make mucus thinner and easier to cough up. One type commonly used by people with CF is called Pulmozyme.
- **Antibiotics:** These may be used to fight off infections that sometimes occur in the lungs of people with CF. There are many types of antibiotics that may be used for people with CF. One type of inhaled antibiotic that is often used for CF treatment is called Tobramycin (TOBI).
- **CFTR modulators:** These drugs enhance or restore the expression, function, and stability of a defective CFTR protein. CFTR modulators are new medicines that can improve the symptoms of CF and may be used in children and adults.
- Other medications may be suggested for children or adults with liver disease, high blood sugar levels, or bone thinning.

Do not use any medication without checking with your doctor.

### **5. Also important for your child's health**

- Have your child vaccinated according to the regular childhood schedule. Children with CF need all the usual childhood vaccinations. It is especially

important for your child to have a measles vaccine. In addition, your doctor may suggest that your child have vaccinations against influenza and pneumonia on a yearly basis. Children with CF should also be protected against RSV, a respiratory illness that can be severe, and sometimes life-threatening, in children with chronic lung disease.

- Keep your child away from all forms of smoke, especially cigarette and cigar smoke. It can add to lung damage.
- Teach good hand washing habits to prevent infection.
- If your child has a respiratory infection and is too sick to eat or follow regular health habits, call your doctor right away. During some illnesses, your child may need to be seen in the hospital for treatment.
- Encourage your child to get plenty of exercise. This will help maintain your child's lung function and improve overall health.

Some adults with CF have severe lung damage that can no longer be treated just with medication. These people have the option of lung transplantation. For more information on CF and lung transplantation see [www.cff.org/treatments/LungTransplantation/](http://www.cff.org/treatments/LungTransplantation/).

You may read information about the potential for gene therapy to treat or cure CF in the future. Researchers are currently trying to find a way to insert a working copy of the CFTR gene into the cells that need it. Although they have not yet found a way to do this safely and effectively, researchers across the country continue to work on gene therapy treatments for CF.

## **What happens when CF is treated?**

There is currently no cure for CF. However, children who receive prompt and careful treatment have the opportunity to live healthier and more productive lives. The goal of treatment is to lessen the health problems that occur with CF.

You may be advised to see a doctor who specializes in caring for children with CF or to be seen at a CF treatment center. These doctors can work with your regular pediatrician to ensure up-to-date treatment for your child.

Although CF is a life-shortening condition, due to the recent discovery of better treatments, more people with CF are living into adulthood and leading healthier lives than in the past.

## **What causes the CFTR protein to be absent or not working correctly?**

Genes tell the body how to make enzymes. The CFTR gene instructs the body to make the CFTR enzyme. Everyone has two copies of the CFTR gene. People with CF have changes, also called variants in both copies of their CFTR genes that cause them to not work correctly. Because of the variants in the CFTR genes, the CFTR enzyme either does not work properly or is not made at all.

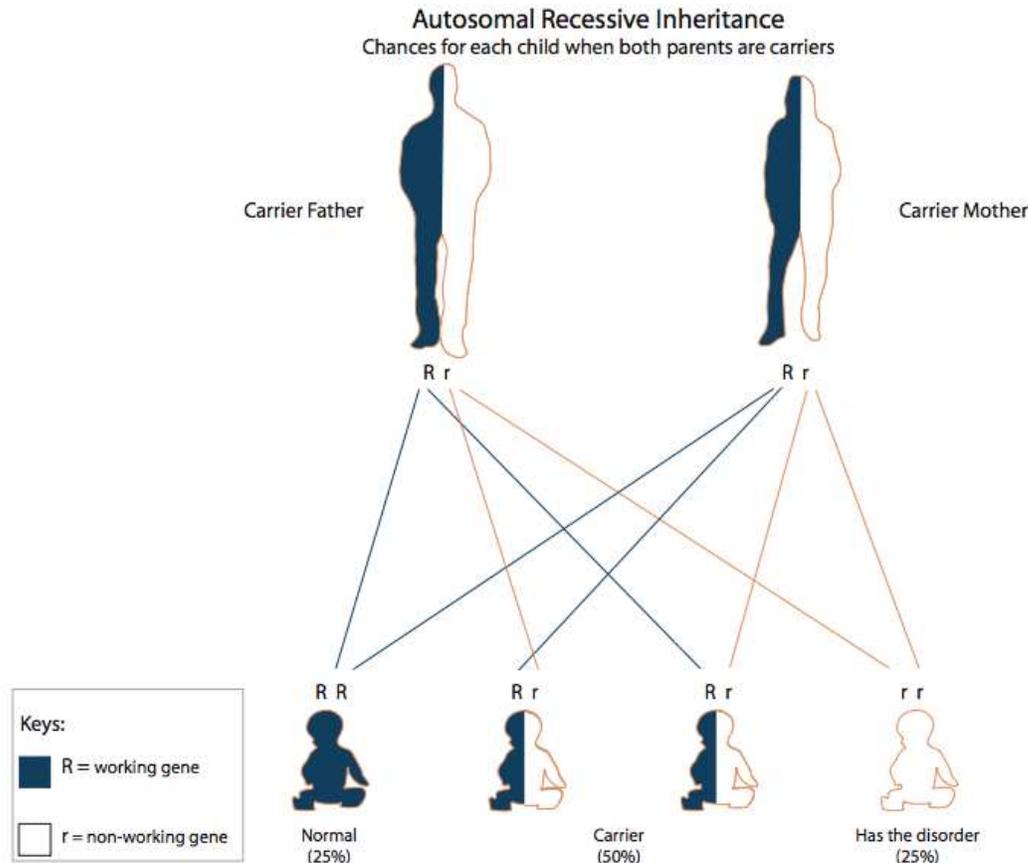
## **How is CF inherited?**

CF is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has two copies of the CFTR genes that make the CFTR protein. In children with CF, neither of these CFTR genes works correctly. These children inherit one non-working gene for the condition from each parent.

Parents of children with CF rarely have the disorder. Instead, each parent has a single non-working CFTR gene for CF. They are called carriers. Carriers do not have CF because their other gene is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CF. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



Genetic counseling is available to families who have children with CF. Genetic counselors can answer your questions about how CF is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

### Is genetic testing available?

Genetic testing for CF can be done either on a blood sample or from a cheek swab. Genetic testing, also called DNA testing, looks for changes (variants) in the pair of genes that cause CF. In most children with CF, both gene changes can be found. However, in other children, only one or neither of the two gene changes can be found, even though we know they are present.

### What other testing is available?

#### **Newborn Screening**

All states offer newborn screening for CF. A blood spot from a needle prick on a baby's heel is used to screen for a number of different genetic conditions. If a baby has a positive result on the initial CF screen, it does not yet mean that he or she has CF. Many babies with a positive result on newborn screening for CF are

later found not to have CF. However, a positive screening test means that further testing must be done to confirm or rule out this condition.

### **Confirmatory testing**

The test used most often to confirm CF is called a 'sweat chloride' test. A small amount of a substance that produces sweat is put onto a small section of a child's arm or leg. A tiny electrical current is used to make that part of the skin sweat. The sweat is collected and the amount of chloride is measured. A high level of chloride in the sweat confirms CF. However, this test does not tell how mild or severe the condition will be.

Sweat chloride tests are often done after a baby is a few weeks old because newborns often don't have enough sweat to do the test properly.

Your child will likely have other medical tests as part of his or her routine care. These may include chest X-rays and other tests to look at the lungs, along with blood and urine tests to make sure the pancreas and liver are working correctly.

### **Can you test during a future pregnancy?**

If both gene changes (variants) have been found in your child with CF, DNA testing can be done during future pregnancies. The sample needed for this test is obtained by either CVS or amniocentesis.

Parents may either choose to have testing during pregnancy or wait until birth to have the baby tested. Parents may also choose to use assisted reproductive techniques to decrease the chance that their future children would have CF. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

### **Can other members of the family have CF or be carriers?**

#### **Having CF**

The brothers and sisters of a baby with CF also have a chance of being affected, even if they have had no symptoms. Finding out whether other children in the family have CF is important because early treatment may prevent more serious health problems. Talk to your doctor or genetic counselor about testing your other children for CF.

#### **CF Carriers**

Brothers and sisters who do not have CF still have a chance to be carriers like their parents. Except in special cases, carrier testing should only be done on people over 18 years of age.

If you are a parent of a child with CF, your brothers and sisters have a 50% chance to be a CF carrier. It is important for other family members to be told that

they could be carriers. There is a small chance they are also at risk to have children with CF.

All states offer newborn screening for CF. However, when both parents are carriers, newborn screening results are not sufficient to rule out CF in a newborn baby. In this case, diagnostic testing should be done in addition to newborn screening.

## **Can other family members be tested?**

### **Diagnostic testing**

If both gene changes have been found in your child with CF, brothers and sisters can be tested for CF using DNA testing on a blood sample or a cheek swab. Other special tests, such as sweat chloride testing may also be suggested.

### **Carrier Testing**

If both gene changes have been found in your child with CF, other family members can have DNA testing on a blood sample or cheek swab to see if they are carriers.

## **How many people have CF?**

About one in every 3,200 white babies in the United States is born with CF. CF is less common in children of other ethnic backgrounds. CF affects about one in 8,000 Latino babies, one in 15,000 African-American babies, and less than one in 30,000 Asian babies.

## **Does CF happen more often in a certain ethnic group?**

CF happens more often in white people from Northern Europe and the United States, but it can affect people of all ethnic backgrounds. About one in every 28 Caucasians in the United States is a CF carrier. In addition, about one in 46 Latinos, one in 65 African-Americans, and one in 90 Asians is a CF carrier.

## **Does CF go by any other names?**

CF is also sometimes called:

- Mucoviscidosis
- Cystic fibrosis of pancreas
- Fibrocystic disease of pancreas

## **Where can I find more information?**

Genetic Alliance

<http://www.geneticalliance.org>

Cystic Fibrosis Foundation (CFF)  
<http://www.cff.org>

Cystic Fibrosis Research Inc. (CFRI)  
<http://www.cfri.org/>

Baby's First Test  
<https://www.babysfirsttest.org/>

Genetic Home Reference  
<https://ghr.nlm.nih.gov/condition/cystic-fibrosis>

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