

Newborn Screening for Cystic Fibrosis

Why is Newborn Screening for Cystic Fibrosis an emerging public health issue?

Cystic Fibrosis (CF) is a genetic disease that affects approximately 1 in 2,500 babies born in the United States and about 1 in 25 Caucasian people are carriers of the CF gene. Currently, there are about 30,000 people in the United States affected by CF. This disorder is characterized by an increased secretion of sticky mucus, which can affect the function of the lungs, pancreas and other organs of the body. Early detection of this genetic disease can help improve the quality of life for those affected by leading to early intervention and treatment of infections and digestive problems. Currently six states are performing newborn screening for CF and continuous studies are being done to evaluate the outcomes and effectiveness of the screen.

Suggested Reading: [Genetic Testing for Cystic Fibrosis: National Institutes of Health Consensus Development Conference Statement](#)

What is Cystic Fibrosis?

CF is a genetic disorder that affects many organs of the body, especially the lungs, pancreas and sweat glands. CF is characterized by the accumulation of abnormal concentrations of salt in the body, which leads to the formation of a thick sticky [mucus](#) that can coat cells. Coated cells in the lungs can lead to the blockage of small airways, which makes it difficult to clear bacteria and can cause infections. Repeated infections can lead to lung damage. This thick mucus can also block the [ducts](#) that carry [digestive enzymes](#) from the [pancreas](#) to the [intestine](#) that help breakdown food. When the enzymes cannot get into the intestine, food is not completely [digested](#). Although a person may seem to consume enough calories to support their body weight, they may lose weight because the body cannot utilize the incompletely digested nutrients in the food. The increased level of salt also leads to an increased level of salt in the individual's sweat.

What are the symptoms of Cystic Fibrosis?

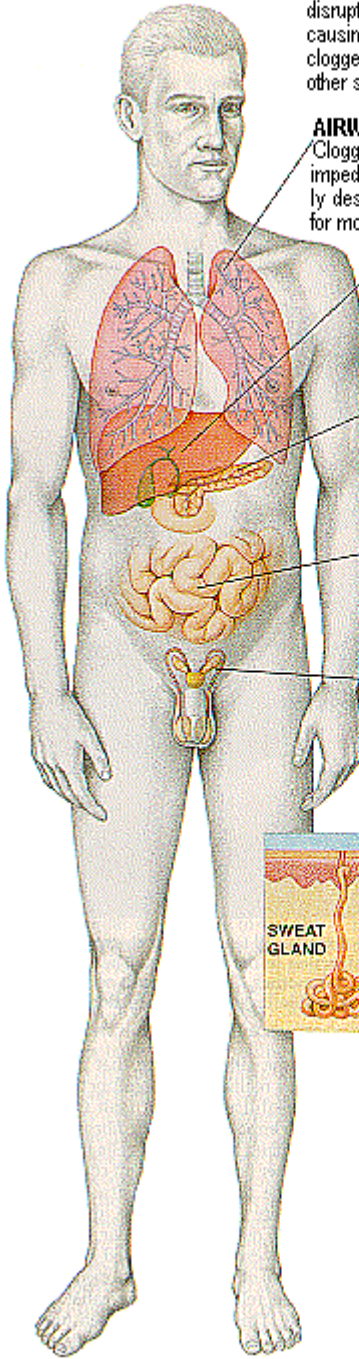
The symptoms of CF vary from individual to individual. There are a number of symptoms that are commonly found amongst individuals, but there are also symptoms that occur much less frequently, such as [infertility](#). Individuals with CF do not have to express each of the common symptoms.

Most Common Symptoms of CF

Salty tasting skin
Constant coughing or wheezing
Frequent & prolonged cases of pneumonia
Excessive appetite
Inability to gain weight
Bulky stools

Organs Affected by Cystic Fibrosis

The genetic defect underlying cystic fibrosis disrupts the functioning of several organs by causing ducts or other tubes to become clogged, usually by thick, sticky mucus or other secretions.



AIRWAYS

Clogging and infection of bronchial passages impede breathing. The infections progressively destroy the lungs. Lung disease accounts for most deaths from cystic fibrosis.

LIVER

Plugging of small bile ducts impedes digestion and disrupts liver function in perhaps 5% of patients.

PANCREAS

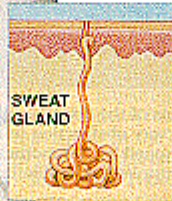
Occlusion of ducts prevents the pancreas from delivering critical digestive enzymes to the bowel in 85% of patients. Diabetes can result as well.

SMALL INTESTINE

Obstruction of the gut by thick stool necessitates surgery in about 10% of newborns.

REPRODUCTIVE TRACT

Absence of fine ducts, such as the vas deferans, renders 95% of males infertile. Occasionally, women are made infertile by a dense plug of mucus that blocks sperm from entering the uterus.



SWEAT GLAND

SKIN

Malfunctioning of sweat glands causes perspiration to contain excessive salt (NaCl). Measurement of chloride in sweat is a mainstay of diagnosis.

ROBERTO-GETTI

What causes cystic fibrosis?

CF is most commonly seen in people of Northern European ancestry. Approximately 1 in 25 Caucasians are [carriers](#) of the CF gene and 1 in 2,500 births in the United States will be affected by CF. Carrier frequency is decreased in people of African, Asian and Hispanic descent.

Although there are more carriers in the Caucasian population, CF can affect people of all races and ethnicities.

CF is an [autosomal recessive](#) disorder caused by a change in a [gene](#) located on [chromosome 7](#). Individuals who carry one gene responsible for CF are called [carriers](#) or [heterozygotes](#). Being a carrier means that you will not have health problems related to CF, but that you have a 50% chance of passing the CF gene onto your children and a 50% chance of passing the normal gene to your children. If your partner is also a carrier of the CF gene he/she has the same chance of passing the CF gene to your children. There is a 25% chance that you will both pass the CF gene to any one of your children. This child would have two copies of the CF gene, known as a [homozygote](#), and develop symptoms characteristic of CF. There is a 50% chance that the child would receive one CF gene and therefore be a carrier like his/her parents. There is also a 25% that the child would not [inherit](#) any copies of the CF gene, so he/she would not develop or be a carrier of CF.

What testing is available for Cystic Fibrosis?

- Newborn screening
- [Sweat Test](#)

Newborn screening consists of a number of tests that can screen infants for certain genetic diseases. Every state in the United States tests for [phenylketonuria](#) (PKU) and [hypothyroidism](#). In addition, each state can opt to perform additional screening tests, including the test for CF. Six states, Colorado, Connecticut, Massachusetts, Montana, Wisconsin, and Wyoming are currently performing a newborn screening tests for CF. Fortunately, most test results come back negative, but screening is very important because early diagnosis and treatment for those affected can help improve the baby's health.

A small blood sample is needed to perform the screen. The baby's heel is pricked to obtain a few drops of blood, which will be used for the various tests. The blood is then sent to the laboratory for analysis. The results will be returned to your child pediatrician.

Although newborn screening has been shown to be highly effective in the identification of infants with CF, the test is not 100% accurate. If your child has a positive test result for CF, he/she will need special medical care throughout his/her life. Your pediatrician may refer you to a geneticist or a genetic counselor to discuss the genetics involved with CF and its related risks. It is important to discuss all of your questions with your doctor. If your child shows the symptoms of CF and the screen was negative, your physician may request that a [sweat test](#) be done to rule out CF. The sweat test is a painless test that measures the amount of salt in the individual's sweat. A high salt concentration is indicative of CF.

There is a blood test that can determine if you are a carrier of CF. You are at an increased risk for being a carrier if there is someone in your family affected with CF or a known carrier of the gene.

The decision to have your child tested for CF is an important decision. Several states include CF in the newborn screening tests that they perform on all children at birth. Recent advances in the treatment of people with CF have led to an increase in the length and quality of life. Currently gene therapy is being studied as a potential treatment for CF and may potentially lead to a cure to this disease. Identifying if your child has CF may be important in making health decisions as

well as reproductive decisions for both parent and child in later life. Talk to your doctor or a genetic counselor about the CF screening tool especially if there is a history of CF in your family.

How is Cystic Fibrosis treated?

Currently there is no cure for CF. Treatment for CF is dependent upon the individual's specific symptoms and the stage that they are at in the disease.

The CF gene was discovered in 1989 and research focusing on a cure for CF, including gene therapy, has been ongoing since that time. More effective medications and treatments for CF are constantly being developed.

Medications

- **Antibiotics:** Antibiotics are used to treat and prevent lung infections and to ease congestion. Unlike antibiotic use in healthy individuals, individuals with CF usually take higher doses of the antibiotic over an extended period of time. They can be administered orally or given [intravenously](#).
- **Bronchodilators:** Bronchodilators, which are commonly used to treat asthma, help to open up and clear out clogged airways by relaxing the muscles around the airways. They are most commonly inhaled, but can also be administered orally or intravenously.
- **Pancreatic [enzyme supplements](#):** can be taken with meals to help in breaking down food.
- **Mucolytics:** Mucolytics are drugs that thin the mucus.
- **Human DNase:** Human DNase is a new therapy that loosens the mucus in the lungs by cleaving DNA in the mucus. This treatment is still considered a supplemental, experimental therapy and is being used only for patients with moderate to severe cases of CF.

Chest physical therapy

- **Percussion:** tapping or “clapping” the chest and back vigorously to loosen the mucus clogging the lungs
- **PEP (positive expiratory pressure) Mask Therapy:** loosens mucus clogging the lungs

Oxygen therapy

Oxygen therapy is used primarily to help prevent heart disease which can be caused by the increased resistance to blood flow through the lungs.

Supplements

A diet rich in [vitamins](#) and [minerals](#) may be recommended to promote good nutrition.

Lung Transplantation

Lung transplantation is an option for individuals with CF. Due to the extremely high cost of the procedure and possible complications that may arise after the procedure, there are specific [criteria](#) used to decide whether a patient with CF should be a candidate for a transplant.

Exercise

Exercise is an effective way to clear the lungs of excess mucus and is essential for maintaining good health and minimizing the complications associated with CF.

Information and Support Resources

- [American Cystic Fibrosis Foundation](#)
- [Canadian Cystic Fibrosis Foundation](#)
- Genetics Education Program of New South Wales, Australia Fact Sheets:
 - <http://www.genetics.com.au/resource/factsht/18.html> (Cystic Fibrosis)
 - <http://www.genetics.com.au/resource/factsht/17.html> (Newborn Screening)
- [March of Dimes](#)
- [National Heart, Lung, and Blood Institute Facts about CF](#)