

Cystic Fibrosis

What is cystic fibrosis?

Cystic fibrosis is an inherited condition of the mucus glands that affects many body systems. The most common signs and symptoms include progressive damage to the respiratory system and chronic digestive system problems. Mucus lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues. In people with cystic fibrosis, the body produces mucus that is abnormally thick and sticky.

About one baby in 2,500-3,500 is born with cystic fibrosis in the United States. The condition can be found in all ethnic groups, but is most common in Caucasian (white) population.

How does cystic fibrosis affect a child?

The thick mucus can obstruct the airways, leading to problems with breathing and infections in the lungs. These infections cause chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections result in permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

In people with cystic fibrosis, mucus blocks the ducts of the pancreas. The pancreas is an organ that produces insulin (a hormone that helps control blood sugar levels). It also makes enzymes that help digest food. This blockage prevents these enzymes from reaching the intestines to aid digestion. Problems with digestion can lead to diarrhea, malnutrition, poor growth, and weight loss.

What causes cystic fibrosis?

Cystic fibrosis is a genetic condition caused by a change in the cystic fibrosis transmembrane conductance regulator (CFTR) [gene](#). Cystic fibrosis is inherited in an [autosomal recessive](#) pattern, which means two copies of the [gene](#) must be changed for a person to be affected with cystic fibrosis. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "[carriers](#)", with one copy of the changed [gene](#) and one copy of the normal [gene](#).

When both parents are [carriers](#), there is a one-in-four (or 25 percent) chance that both will pass the changed [gene](#) on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal [gene](#), and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed [gene](#) from one parent and a normal [gene](#) from the other, making it a [carrier](#) like its parents. These chances are the same in each pregnancy with the same parents.

Is there a test for cystic fibrosis?

Yes. Babies are tested (newborn screening) for cystic fibrosis before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to look at the immunoreactive trypsinogen (IRT).

Can cystic fibrosis symptoms be prevented?

The types of treatment your child receives depends on what kinds of health problems the cystic fibrosis is causing and how your child's body responds to different types of treatment. Most people combine medicines, home treatment methods (breathing exercises), high-energy foods, enzyme supplements, and vitamin and mineral supplements. Regular exercise is important to keep the body fit and healthy. Consults with your pulmonologist and /or regional genetic center to determine what tests and follow up are needed.

DISCLAIMER: The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

Resources:

Cystic Fibrosis Foundation (national headquarters)
6931 Arlington Road
Bethesda, Maryland 20814
Phone: Toll free: (800) FIGHT CF (344-4823)
Fax: 1-301-951-6378
E-mail: info@cff.org

MUMS National Parent-to-Parent Network
Julie J. Gordon
150 Custer Court
Green Bay, Wisconsin 54301-1243
Phone: 1-877-336-5333 (Parents only please)
Phone: 1-920-336-5333
Fax: 1-920-339-0995
E-mail: mums@netnet.net
www.netnet.net/mums/

References:

- Genetics Home Reference
<http://ghr.nlm.nih.gov/>
- Online Mendelian Inheritance in Man (OMIM topics 219700)
<http://www.ncbi.nlm.nih.gov/Omim>