

What is cystic fibrosis?

Cystic fibrosis is an inherited disease that in its classic form is characterized by progressive lung damage, chronic digestive problems, decreased pancreatic function, and male infertility. Individuals with cystic fibrosis have abnormalities in a protein, the cystic fibrosis transmembrane conductance regulator (CFTR), which controls the movement of chloride (salt) and water across cell membranes, resulting in excess salt in the sweat glands and the production of abnormally thick mucus. The symptoms of cystic fibrosis are due to the thickened mucus that clogs the airways and various glands in the body.¹

What are the symptoms of cystic fibrosis and what treatment is available?

Cystic fibrosis is a disease that varies in severity and age at presentation, even within families. Signs of cystic fibrosis may be visible by prenatal ultrasound but typically occur in infancy or childhood. Signs and symptoms of the classic form of cystic fibrosis include lung, pancreatic, and gastrointestinal symptoms, infertility and a reduced life expectancy. Some individuals may experience fewer or milder symptoms, which can include isolated male infertility.² Some individuals may reach adulthood and not be aware that they have the disease.³

Symptoms of cystic fibrosis may include:^{1,2,4}

- Meconium ileus (intestinal blockage due to abnormally thick fetal stool) visible on prenatal ultrasound or detected at birth
- Salty tasting skin
- Chronic sinus and lung disease (coughing, wheezing, sinusitis, and frequent infections)
- Poor growth and weight gain
- Pancreatic insufficiency (causing diarrhea and a reduced ability to digest and absorb nutrients from food)
- Pancreatitis (inflammation of the pancreas)
- Diabetes
- Infertility in males due to congenital absence of the vas deferens
- Distal intestinal obstruction syndrome (intestinal blockage due to undigested material)
- Liver disease

There is no cure for cystic fibrosis. Treatment may include respiratory therapy, antibiotics, and nutritional and pancreatic enzyme supplementation. New medications are becoming available that treat the underlying protein defect. In severely affected individuals, lung transplantation may be considered.² The median life expectancy for individuals with classic cystic fibrosis is about 40 years.⁵

Cystic fibrosis is included on all newborn screening panels in the United States.⁶

How is cystic fibrosis inherited?

Cystic fibrosis is an autosomal recessive disease caused by mutations in the *CFTR* gene.¹ An individual who inherits one copy of a *CFTR* gene mutation is a carrier and does not usually have related health problems. An individual who inherits two *CFTR* mutations, one from each parent, is expected to be affected.

If both members of a couple are carriers, the risk for having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for cystic fibrosis?

Cystic fibrosis can occur in individuals of all races and ethnicities, but it is most common in individuals of Ashkenazi (Eastern European) Jewish and Caucasian ancestry.³ In the United States, the incidence is approximately 1 in 3500.⁵

Estimated carrier frequency for select ethnic groups³

Population	Carrier frequency
Ashkenazi Jewish	1 in 24
Caucasian	1 in 25
Hispanic	1 in 58
African American	1 in 61
Asian American	1 in 94

Having a relative who is a carrier or who is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Cystic fibrosis.com: www.cysticfibrosis.com

Cystic Fibrosis Foundation: www.cff.org

Cystic Fibrosis Research Inc.: <http://www.cfri.org/home.shtml>

References

1. Cystic Fibrosis Genetics Home Reference Available at: <http://ghr.nlm.nih.gov/condition/cystic-fibrosis>. Accessed on: March 14, 2012.
2. Moskowitz SM *et al.* Cystic fibrosis. *Gene Reviews*. Available at: <http://www.ncbi.nlm.nih.gov/books/NBK1250/>. Accessed: Mar 10, 2012.
3. Update on carrier screening for cystic fibrosis. ACOG Committee Opinion Number 486. American College of Obstetrics and Gynecologists. *Obstet Gynecol* 2011; 117:1028-1031.
4. Zielenski J. Genotype and phenotype in cystic fibrosis. *Respiration*. 2000; 67:117-133.
5. Cystic Fibrosis Patient Registry 2013 Annual Data Report. Cystic Fibrosis Foundation.
6. National Newborn Screening Status Report. November 2, 2014. Available at: <http://genes-r-us.uthscsa.edu/sites/genes-r-us/files/nbsdisorders.pdf>. Accessed January 6, 2016.