

Cystic Fibrosis

Cystic fibrosis (CF) is a **genetic** (inherited) disease that affects mainly the lungs, the pancreas, and the sweat glands. Some CF patients develop serious liver disease. The abnormal CF gene changes the makeup of mucus in the airways, making it thick and sticky. The body cannot clear this thick mucus from the airways, leading to recurrent lung infections that are difficult to treat. Thick secretions also block other glands, causing them to function poorly. About 30 000 persons in the United States have CF and more than 10 million carry the gene for CF. Carriers of the gene do not have symptoms of CF, but if they have a child who inherits 2 copies (one from each biological parent) of the CF gene, that child will develop cystic fibrosis and can also pass the CF gene on to any children he or she may have. In the past, most persons with CF found out about their condition when they had symptoms in childhood. Newborn screening programs can now lead to earlier diagnosis and therapy to limit the impact of the disease. The September 9, 2009, issue of *JAMA* includes an article about genetic variations that contribute to the development of serious liver disease in cystic fibrosis.

SIGNS AND SYMPTOMS

- Frequent lung and sinus infections
- Greasy frequent stools, often with abdominal pain and bloating
- Malnutrition and poor weight gain in infants

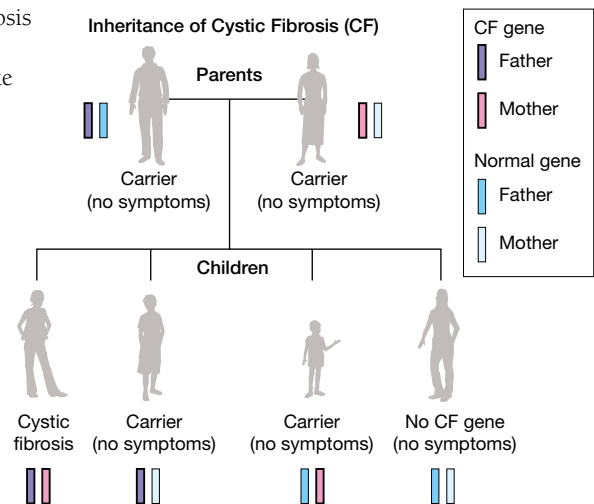
DIAGNOSIS AND TESTING

The most common test for CF is the sweat test, in which the salt (sodium chloride) content of the sweat is measured. Persons with CF have high concentrations of salt in their sweat because of abnormal sweat gland function. The gene for CF was identified in 1989 and genetic testing is now available. This can be done with **prenatal** (before birth) testing or in infants, children, and adults. Carriers of the CF gene can also be identified with genetic testing, which is offered as a component of family planning.

TREATMENT

- Persons who have CF are usually cared for by a team of specialists with expertise in this disease. Treatment is specific to each problem caused by CF. Breathing treatments, chest physiotherapy (specialized physical therapy to encourage drainage of lung secretions), exercise programs, and antibiotics may be used to treat lung disease and infections. Oxygen may be used for more severe lung disease due to CF.
- Replacement of **pancreatic enzymes** (substances made by the pancreas to help healthy digestion) is typically needed. Multivitamins are usually prescribed because of potential vitamin deficiency due to poor absorption in the intestine.
- For severe lung disease, placement of long-term intravenous (IV) catheters or feeding tubes may be required. Lung transplantation may be an option to improve functional status for some persons with severe CF, but transplantation does not cure CF and has risks of its own.
- There is no cure for CF. However, treatment of CF and its complications has improved so much in the last 50 years that survival and quality of life for children and adults with CF is much better than it was in the past. The median age of survival for persons with CF is now more than 37 years, double what it was 25 years ago.

Inheritance of Cystic Fibrosis (CF)



FOR MORE INFORMATION

- Cystic Fibrosis Foundation
www.cff.org
- American Lung Association
www.lungusa.org
- National Heart, Lung, and Blood Institute
www.nhlbi.nih.gov

INFORM YOURSELF

To find this and previous JAMA Patient Pages, go to the Patient Page link on JAMA's Web site at www.jama.com. Many are available in English and Spanish.

Sources: American Lung Association; National Heart, Lung, and Blood Institute; Cystic Fibrosis Foundation

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