

## Twenty Facts About Cystic Fibrosis

Cystic Fibrosis is a progressive genetic disease that affects the lungs and other body systems and for which there is currently no cure.



1. Cystic fibrosis (CF) is the most common fatal genetic (inherited) disease in North America.
2. CF occurs when a person inherits a mutated (abnormal) copy of the CFTR (cystic fibrosis transmembrane conductance regulator) gene from each parent. Approximately 2000 CFTR gene mutations have been linked to disease.
3. CF is inherited as an autosomal recessive disease, meaning only people with 2 CFTR mutations have the disease. People with only one CFTR mutation are carriers and do not have the disease. If both parents are carriers, there is a 1 in 4 chance that their child will receive an abnormal CFTR gene from each parent and have the disease.
4. While CF is more common in Caucasians, CF occurs in all races and ethnicities.
5. While there is no cure, life expectancy has steadily improved with the median survival exceeding 45 years in the United States. However, the median age of death is approximately 30 years. The median is the age that separates the older from younger halves of the group who live or those who die from CF. Half of those who die do so by or before 30 years.
6. There are now more adults than children with CF in the United States.
7. The defective gene causes the CFTR protein to not form or work properly, causing abnormal movement of salt and water across the cell, which leads to dehydration of the airway surface and thick mucus that clogs ducts throughout the body.
8. The sweat chloride measurement, or sweat test, is the recommended test to diagnose CF, since affected people have higher sodium and chloride levels, making the sweat more salty.
9. Newborn screening for CF done on blood samples can identify most children before one month of age, which allows for early treatment and disease monitoring.
10. Older children and adults are usually diagnosed based on symptoms, such as frequent respiratory infections, malnutrition, and/or male infertility.
11. New medications can now help improve CFTR function in the cells of some patients based on the type of gene mutations they have.
12. CF individuals have abnormally thick mucus, which blocks the airways (obstruction) and leads to repeated infections and damaging inflammation in the lungs. Treatments are directed at trying to prevent and treat these problems.

13. Infection control is important to help avoid exposure to dangerous pathogens (germs including bacteria and viruses that cause illness). People with CF are discouraged from close contact with other CF patients, and should try to limit exposure to certain bacteria and viruses.
14. Immunizations, including a yearly flu shot, are important preventive measures and recommended for CF patients and caregivers.
15. Due to the thick mucus blocking the ducts in the pancreas and preventing the release of pancreatic enzymes that help break down fat and protein, most people with CF suffer from malabsorption of food, leading to unhealthy low weight and nutritional issues. In addition to needing to enzyme replacement, they need to take extra fat-soluble vitamins (A, D, E, and K) and calories to stay healthy.
16. The thick mucus also blocks ducts in other organs, causing additional health problems including liver disease, CF related diabetes and pancreatic insufficiency. Other health issues can develop in CF such as sinus disease, osteoporosis, and reproductive health issues. The CF health care team helps monitor and manage them.
17. CF care requires a team that includes the individual's family, a primary care doctor, as well as CF doctors, nurses, dietitians, social workers, pharmacists, respiratory therapists, and others. There is a network of specialized care centers in the United States that are accredited by the Cystic Fibrosis Foundation (CFF) offering patients high quality care. To find a care center, contact the Cystic Fibrosis Foundation website ([www.cff.org/Care-Centers/Find-a-CF-Care-Center](http://www.cff.org/Care-Centers/Find-a-CF-Care-Center)).
18. Regular monitoring of lung function tests and sputum cultures helps the health care team advise individuals with CF on how to keep their lungs healthy and adjust treatments with any change. The CFF recommends that all patients with CF be seen in a specialized care center at least every 3 months.
19. Resources are available to help people with CF struggling with insurance, health care, school or other life or work issues. Contact the CFF Compass Program ([www.cff.org](http://www.cff.org)), Cystic Fibrosis Research, Inc. ([www.cfri.org](http://www.cfri.org)), or ask your CF health care team for help.
20. Good adherence with home therapies as they are prescribed is a key to living as healthy and long a life as possible. Individuals that have trouble doing their home treatment plan should talk to their health care team.

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### Other Resources:

**Cystic Fibrosis Foundation**  
[www.cff.org](http://www.cff.org)

**American Thoracic Society**  
[www.thoracic.org/patients](http://www.thoracic.org/patients)

**Cystic Fibrosis Research, Inc.—  
an American Thoracic Society PAR organization**  
[www.cfri.org](http://www.cfri.org)

**European Cystic Fibrosis Society (ECFS)**  
[www.ECFS.org](http://www.ECFS.org)

**Cystic Fibrosis Trust (United Kingdom)**  
[www.cysticfibrosis.org.uk](http://www.cysticfibrosis.org.uk)

**Boomer Esiason Foundation**  
[www.esiason.org](http://www.esiason.org)

**United States National Library of Medicine**  
[www.medlineplus.gov/cysticfibrosis.html](http://www.medlineplus.gov/cysticfibrosis.html)

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