What Is Cystic Fibrosis?

Cystic fibrosis (CF) is a genetic disorder caused by an abnormal gene that is inherited from both biological parents.

The cystic fibrosis transmembrane conductance regulator (CFTR) gene is responsible for salt transport across different tissues in the body. In CF, the protein made by the abnormal CFTR gene is absent or dysfunctional, resulting in reduced salt transport and decreased water movement, which causes thick mucus to accumulate in various parts of the body.

How Common Is Cystic Fibrosis?
Cystic fibrosis affects more than 30,000 children and adults in the US and 70,000 people worldwide. Although CF affects people of all races and ethnicities, it is most common in non-Hispanic White individuals.

Diagnosis of Cystic Fibrosis
Newborn screening programs measure a chemical in the blood called immunoreactive trypsinogen, which is elevated in CF. Most newborn screening programs also include genetic testing for the most common mutations that cause CF. Diagnosis is confirmed by finding elevated chloride levels in sweat. The sweat chloride test is also used for diagnosis of CF if the condition is clinically suspected in a child or adult.

How Does Cystic Fibrosis Affect Body Functions?
Abnormal or absent CFTR protein causes accumulation of thick mucus, which blocks the airways, leads to repeated episodes of infection and inflammation, and results in damage to the lungs. Respiratory failure is the most common cause of death in people with CF.

People with CF also have thick mucus in the pancreas, which limits release of digestive enzymes and leads to difficulty digesting food, malabsorption of nutrients, and poor weight gain. In addition, damage to the pancreas can lead to development of CF-related diabetes. People with CF may have chronic diarrhea and episodes of constipation.

Other parts of the body can also be affected. Most men with CF are infertile, and women with CF may have difficulty becoming pregnant. People with CF often have chronic sinusitis. Also, people with CF are at increased risk of dehydration in hot weather because of abnormal functioning of their sweat glands.

Treatments for Cystic Fibrosis
Most people with CF take inhaled medications daily to thin their mucus and use mechanical devices several times daily to dislodge mucus from the airways. Oral and inhaled antibiotics may be prescribed to help control infection, while intravenous antibiotics are used to treat flares of infection.

CFTR modulators, the first class of drugs aimed at treating the underlying cause of CF, became available in 2011. CFTR modulators have effects on abnormal CFTR proteins within cells and improve salt transport. In the US, an estimated 90% of people with CF have CFTR gene mutations that may respond to CFTR modulators. These drugs improve lung function, weight gain, and quality of life; decrease episodes of infection; and are expected to improve survival. Among children born with CF in 2019, half are predicted to live to age 48 years or older, an increase of about 10 years since 2009.

A double lung transplant may be an option for some patients with advanced lung disease due to CF.

For More Information
Cystic Fibrosis Foundation
www.cff.org/intro-cf/about-cystic-fibrosis

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