Cystic Fibrosis: Causes, Symptoms, and Treatment

Highlights

Cystic fibrosis \(CF\) is a disease that primarily affects the lungs and digestive system.
CF is caused by a mutation \(change\) in a gene that is inherited from both parents.
There is currently no cure for CF.

In This Article

What is Cystic Fibrosis?

Cystic fibrosis \(CF\) is a genetic disease that primarily affects the lungs

and digestive system, resulting from a change in a specific gene. CF causes thick and sticky mucus to accumulate in the lungs, airways, and digestive system, leading to problems with digestion and repeated lung infections, which can permanently damage the lungs.

What Are the Symptoms of Cystic Fibrosis?

Symptoms of CF can vary from person to person. Some of the common signs include:

Chronic cough
Wheezing
Shortness of breath
Frequent lung and sinus infections

Digestive issues are also common due to improper functioning of the pancreas, leading to symptoms such as:

Poor growth
Difficulty gaining weight
Frequent greasy or oily stools
Bowel blockages

Other possible problems include:

Fertility issues
CF-related diabetes
Incontinence
Reflux
Low bone mineral density

What Causes Cystic Fibrosis?

CF is caused by a mutation in the cystic fibrosis transmembrane conductance regulator \(CFTR\) gene, which controls the balance of salt and water in cells. This gene affects the production of mucus, sweat, and digestive fluids.

Both parents must carry the CF gene for their child to inherit the condition. If both parents carry the gene, the chances of passing on CF are:

1 in 4 chance the child will have CF
1 in 4 chance the child will not have CF
2 in 4 chance the child will be a carrier of the CF gene

How is Cystic Fibrosis Diagnosed?

CF is often diagnosed in newborns through a heel prick test, which checks for several conditions, including CF. If the test is positive, a sweat test is performed at around 6 weeks to confirm the diagnosis.

How is Cystic Fibrosis Treated?

CF is managed by a multidisciplinary team that may include:

Doctors
Nurses
Dietitians
Physiotherapists
Pharmacists
Social workers
Psychologists

Treatment typically includes:

Daily physiotherapy to clear the lungs
Enzyme tablets to aid digestion
Antibiotics for lung infections
Puffers to open the airways
Salt and vitamin supplements
A high-calorie, high-salt, and high-fat diet
Regular exercise to build core strength

Medications called ’modulator therapies’ are now available to target the underlying cause of CF. Some individuals with CF may require organ transplants, such as lung, liver, or pancreas transplants, to prolong their lives.

Complications of Cystic Fibrosis

As life expectancy increases, people with CF are more likely to develop chronic health conditions associated with aging, such as:

Colorectal cancer
Cardiovascular \(heart\) disease

It is not yet clear how modulator therapy will impact the development of these chronic conditions.

Tools And Assistance

People with CF are generally advised not to socialize in person with others who have CF to avoid the risk of cross-infection. Support and services for individuals with CF, their carers, and families are available through various organizations. You can also contact a 24-hour helpline for immediate support and advice.