

# Understanding Cystic Fibrosis Clinical Symptoms and Disease Manifestations

Education about cystic fibrosis (CF) to better understand the complex medical profiles of individuals with the disease.

CF is a systemic disease that may result in clinical manifestations throughout the body<sup>1</sup>

CF is a progressive genetic disease that affects the lungs, pancreas, and other organs.

CF is caused by mutations in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene, which results in dysfunctional CFTR protein and leads to thick, sticky mucus in many organs.

- For example, in the lungs, the mucus clogs airways and can trap bacteria, leading to infection, inflammation, and other complications

## Key clinical manifestations of CF<sup>2</sup>

### Pulmonary symptoms

- Frequent lung infections
- Inflammation
- Reduced lung function
- Progressive lung disease

### Pancreatic insufficiency

Exocrine dysfunction

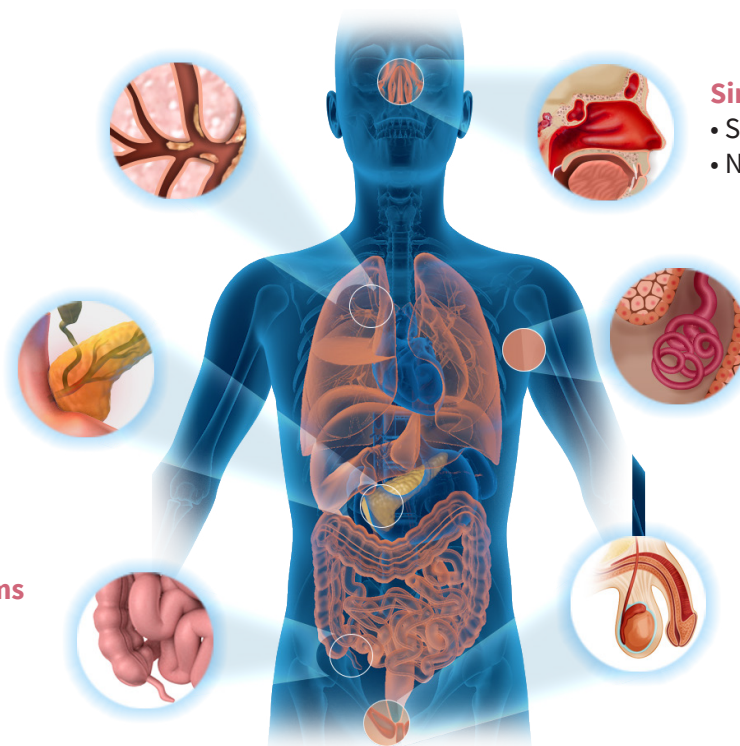
- Malnutrition
- Failure to thrive
- Difficulty with weight gain

Endocrine dysfunction

- Cystic fibrosis-related diabetes

### Gastrointestinal symptoms

- Constipation
- Bloating
- Abdominal pain
- Intestinal blockages



### Sinus symptoms

- Sinusitis (infection)
- Nasal polyps

### Glandular malfunction

- Abnormally high concentration of chloride in the sweat

### Male reproductive dysfunction

- Infertility due to congenital bilateral absence of the vas deferens in men

## Disease manifestations and complications associated with or exacerbated by CF<sup>2,3</sup>



### In the lungs

- Chronic inflammation or obstruction of the airways causes bronchiectasis
- Pulmonary exacerbations caused by airway inflammation and blockage in the airways and lungs
- Collapsed lung, lung infections, and lung damage that may lead to heart failure



### In the gastrointestinal tract

- Meconium ileus
- Distal intestinal obstruction syndrome
- Chronic constipation



### In other body systems

- Increased risk of cancers of the digestive tract: esophagus, stomach, bowels, liver, and pancreas
- Fertility problems
- Mental health problems, such as depression and anxiety
- Muscle and bone complications
- Salt loss syndrome
- Urinary incontinence



### In the pancreas

- Pancreatic insufficiency
- Pancreatitis and low levels of pancreatic enzymes lead to malnutrition, nutritional deficiencies, and low levels of vitamins A, D, E, and K
- Damage to the pancreas can lead to CF-related diabetes



### In the liver

- Blocked bile ducts can lead to liver damage
- Liver damage can advance to cirrhosis
- Cirrhosis may call for a liver transplant

## CF disease progressively worsens over time<sup>2</sup>

The clinical manifestations of CF affect multiple organ systems. Different signs, symptoms, or complications tend to manifest separately or together at different ages. All manifestations stem from defects in the CFTR protein. Examples of CF progression are below.

## Progression of CF disease symptoms and manifestations over time<sup>2</sup>

Infancy	Childhood	Adolescence/adulthood
<b>Lungs</b>		
Lung infection and inflammation	Allergic bronchopulmonary aspergillosis, a heightened reaction to fungi in the environment	Lung damage that may lead to heart failure
<b>Pancreas</b>		
Obstruction of pancreatic ducts and pancreatic insufficiency	Chronic pancreatitis	CF-related diabetes
<b>Liver</b>		
Blocked bile ducts	Liver disease	Liver failure, potentially a need for a liver transplant
<b>Gastrointestinal system</b>		
Meconium ileus (bowel obstruction)	Distal intestinal obstruction syndrome	Distal intestinal obstruction syndrome

**References:** 1. Cystic Fibrosis Foundation. About cystic fibrosis. <https://www.cff.org/What-is-CF/About-Cystic-Fibrosis>. Accessed March 3, 2022. 2. O'Sullivan BP, Freedman SD. Cystic fibrosis. *Lancet*. 2009;373(9678):1891-1904. 3. National Heart, Lung, and Blood Institute. Cystic fibrosis. <https://www.nhlbi.nih.gov/health-topics/cystic-fibrosis>. Accessed March 3, 2022.

