# Cystic Fibrosis Overview

**Cystic fibrosis is a complex lifelong disease.[1] For care managers to better understand the disease, its cause, and the value of a cystic fibrosis care team.**

## What is cystic fibrosis?

Cystic fibrosis (CF) is a progressive, life-shortening, genetic disease[2] that occurs in people who inherit two defective copies of the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene, one from each parent.[3] Mutations in the *CFTR* gene can disrupt the normal production and/or function of the CFTR protein, leading to a buildup of thick fluids that are poorly cleared from passageways in the respiratory tract, digestive system, pancreas, liver, and other organs.[3]

**CF is a rare disease with no cure affecting[1,4,5]:**

- ~ 70,000 people **worldwide**
- More than 30,000 people in the United States, and occurs more commonly in the Caucasian population than in Asian Americans and African Americans

Due to treatment advances, the expected lifespan of people with CF has grown. The median predicted survival age for people with CF born in 2020 is about **59 years old**, which is 9 years longer than the median survival of 50 years for those born between 2016 to 2020.[4]

## Key clinical manifestations of CF[1]

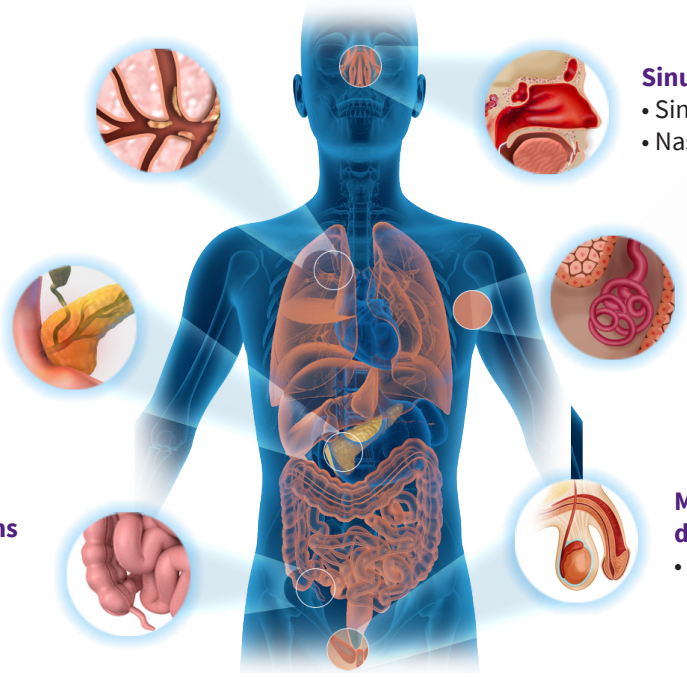
As a multisystemic disease, CF may present with many clinical manifestations in various body systems, as displayed in this illustration.

**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 (CFRD)

**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 (CBAVD) in men

## Symptoms of CF[6]

Some people with CF may have fewer signs or symptoms, yet others experience severe symptoms or life-threatening complications. Symptoms of CF depend on which organs are affected and the severity of the condition.

### CF disease manifestations or symptoms may include[6]:

- **Pulmonary problems**, including serious lung infections
- **Blockage** of the intestine in a baby soon after birth
- **Gastrointestinal symptoms**, such as severe abdominal pain, chronic diarrhea, or constipation
- Low body mass index **(BMI)** or being underweight
- **Salty skin** and saltier-than-normal sweat
- **Sinus** infections

## Milestones in living with CF[3,7,8]

**Children with CF**
grow up, go to school, have friends, have hobbies, and can exercise and play sports

**Adolescents with CF**
work with CF care teams to assume many CF-related responsibilities from parents

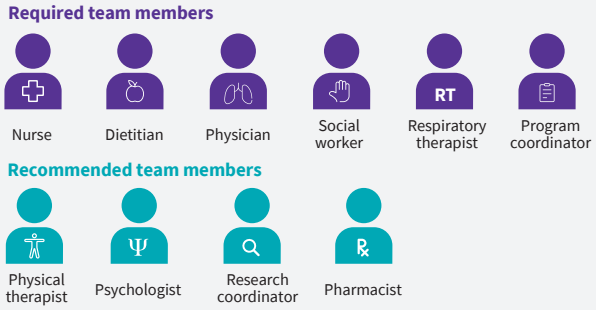
**Young adults with CF**
learn about their treatments and interact independently with CF clinics

**Adults with CF**
manage issues such as independent living, marriage, family planning, and careers

## CF is a complex and chronic disease requiring an expert CF care team[8]

People with CF do best when care is comprehensive, coordinated, and done in partnership with their CF care teams. A team approach ensures that people with CF can receive high-quality, specialized care.

### The multidisciplinary team[8]




**Patients with CF work with their care teams to manage all aspects of their disease**

**References: 1.** O'Sullivan BP, Freedman SD. Cystic fibrosis. *Lancet*. 2009;373(9678):1891-1904. **2.** Cutting GR. Cystic fibrosis genetics: from molecular understanding to clinical application. *Nat Rev Genet*. 2015;16(1):45-56. **3.** Cystic Fibrosis Foundation. CF genetics: the basics. https://www.cff.org/intro-cf/cf-genetics-basics. Accessed March 3, 2022. **4.** Cystic Fibrosis Foundation. 2020 Annual Data Report. https://www.cff.org/about-us/annual-report. Accessed March 3, 2022. **5.** American Lung Association. Learn about cystic fibrosis. http://www.lung.org/lung-health-and-diseases/lung-disease-lookup/cystic-fibrosis/learn-about-cystic-fibrosis.html. Accessed March 3, 2022. **6.** National Heart, Lung, and Blood Institute. Cystic fibrosis. https://www.nhlbi.nih.gov/health-topics/cystic-fibrosis. Accessed March 3, 2022. **7.** Cystic Fibrosis Foundation. Parent and guardian guidance. https://www.cff.org/parent-and-guardian-guidance. Accessed March 3, 2022. **8.** Cystic Fibrosis Foundation. Your CF care team. https://www.cff.org/Care/Your-CF-Care-Team. Accessed March 3, 2022.

VERTEX