Maxor SPECIALTY PHARMACY

Rare Lung Disease
Cystic Fibrosis

Disease State Overview

Cystic Fibrosis (CF) is a rare, genetic disorder that is passed from parents to children. It is caused by inheriting two defective (or abnormal) copies of the gene cystic fibrosis transmembrane conductance regulator (CFTR) protein. This mutation makes mucus and other secretions much thicker and difficult to clear. As a result, there is a build-up of mucus and thick fluids, which creates blockage and obstructs vital organs. CF is a multi-systemic disease that affects the respiratory tract, pancreas, digestive system, and liver. Doctors can screen for and diagnose CF at birth with a positive newborn screening test. Diagnosis is further confirmed with a pilocarpine-induced sweat test and genetic testing. More than 30,000 people in the U.S. are living with cystic fibrosis and around 1,000 people are diagnosed with CF each year.

Related Symptoms and Health Concerns

People experience different CF symptoms because CF affects multiple organs:

- **Lungs**: chronic inflammation and obstruction, lung infections, and trouble breathing. Common symptoms include coughing up thick mucus, wheezing, difficulty breathing, frequent lung infections, and airway inflammation.
- **Pancreas**: low enzyme and vitamin levels needed for absorbing nutrients. Damage can lead to CF-related diabetes.
- **Liver**: blocked bile ducts, which can cause damage to the liver.
- **GI tract**: constipation or obstruction in the intestines. Symptoms include abdominal pain, constipation, malnutrition, or inadequate weight gain.
- **Miscellaneous**: fertility problems, urinary incontinence, and increased risk of GI-related cancers.

Treatment

CF is treatable, but it does not have a cure. CF is more manageable with newer drugs coming to market.

- Air clearance: Loosens mucus build up in the lungs.
- Inhaled medications: Thins mucus and opens airways.

- Oral medications:
  - Pancreatic enzymes to increase absorption of necessary nutrients.
  - CFTR modulators that target and improve function of defective protein.
  - Anti-inflammatory to reduce inflammation.
  - Antibiotics to help treat or prevent infection and inflammation.
  - Nutritional supplements with high dose fat-soluble vitamins.

Treatment Goals

The primary goal of cystic fibrosis treatment is to reduce symptom severity, slow disease progression, and increase survival by...

- Maintaining lung function
- Preventing and treating lung infections
- Loosening and removing inflammation-causing mucus from the lungs
- Preventing and treating of blockages in the intestines
- Promoting adequate nutrition to maintain growth and appropriate BMI
- Encouraging adequate hydration
- Improving or maintaining quality of life
- Increasing survival and life expectancy
- Maintaining optimal therapy adherence
- Preventing, minimizing, and managing side effects

Strategies to Achieve Treatment Goals

- Adhere to drug therapy
- Improve diet and nutrition to maintain growth and appropriate BMI
- Prevent, minimize, and manage side effects
- Prevent and treat lung infections
- Minimize disruption to daily life due to CF symptoms and treatment burden

Additional Resources

- Cystic Fibrosis | CDC | https://www.cdc.gov/genomics/disease/cystic_fibrosis.htm
- Cystic Fibrosis Foundation | https://www.cff.org/
- American Lung Association | Cystic Fibrosis | https://www.lung.org/lung-health-diseases/lung-disease-lookup/cystic-fibrosis
- Commission for Case Manager Certification | Care Management Toolkits | Cystic Fibrosis | https://www.caremanagementtools.com/toolkit/cystic-fibrosis

Sources

1. Lahiri, Thomas, et al. "Clinical Practice Guidelines From the Cystic Fibrosis Foundation for Preschoolers With Cystic Fibrosis." American Academy of Pediatrics, American Academy of Pediatrics, 23 Mar. 2016, pediatrics.aappublications.org/content/early/2016/03/22/peds.2015-1784.
2. Mogayzel, Peter J, et al. "Cystic Fibrosis Pulmonary Guidelines Chronic Medications for Maintenance of Lung Health." *American Journal of Respiratory and Critical Care Medicine*, 3 Jan. 2013, www.atsjournals.org/doi/full/10.1164/rccm.201207-1160OE.