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### Cystic Fibrosis

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# CYSTIC FIBROSIS

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## Overview

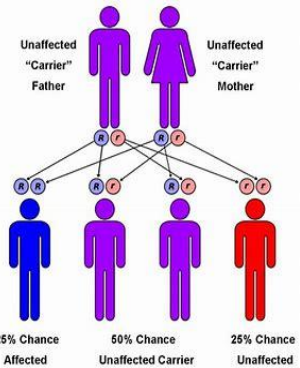
Cystic fibrosis (CF) is one of the lethal autosomal recessive disease that "causes persistent lung infections and limits the ability to breathe overtime" (Cystic Fibrosis foundation, 2018).

### Facts about Cystic Fibrosis

- ✓ Life Expectancy of people with CF is mid- to late 30s
- ✓ More than 30, 000 of people live with CF ( 70,000 worldwide)
- ✓ Approximately, 1,000 new cases are diagnosed each year
- ✓ More than 75 % of people with CF are diagnosed by age 2
- ✓ More than half of CF population is 18 and older (Cystic Fibrosis Foundation, 2018).
- ✓ Lungs disease are 80% of CF mortality (Cutting, 2015, p.45)

### Risk factors

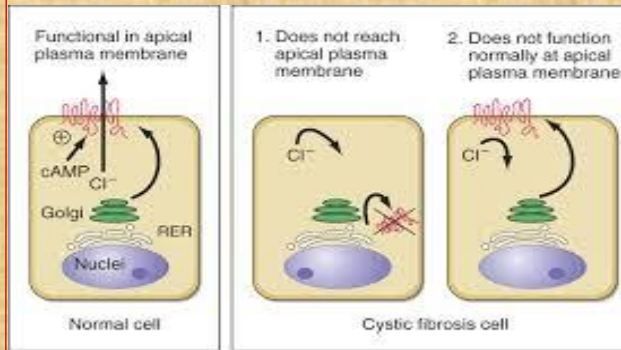
- ✓ Race: commonly in white of Northern Europe (but occurs in all races and ethnicities)
- ✓ Males and females
- ✓ More than 10 million Americans are carriers of a faulty CF gene (NIH, 2018).
- ✓ Family history: Inherited disorder, both parents must have a least one copy of the defective gene for the child to have the disease or (Cystic Fibrosis Foundation, 2018)



## Pathophysiology

CF affects the cells that produce mucus, sweat and digestive juices. These secreted fluids are normally thin and slippery. But in people with CF, a defective gene causes the secretions to become sticky and thick. Instead of acting as a lubricant, the secretions plug up tubes, ducts and passageways especially in the lungs and pancreas (Mayo Clinic, 2016).

CF is caused by defects in the cystic fibrosis gene, which codes for a protein transmembrane conductance regulator (CFTR) that functions as a chloride channel and is regulated by cyclic adenosine monophosphate (cAMP). Mutations in the CFTR gene result in abnormalities of cAMP-regulated chloride transport across epithelial cells



**Table 1**  
Summary of different classes of CFTR mutations

Mutation Class	Nature of defect	Functional consequence
I	CFTR protein synthesis	Reduced CFTR protein expression
II	CFTR protein processing	Misfolded CFTR not transported at to cell surface
III	CFTR channel gating	Reduced/lack of CFTR channel opening
IV	CFTR channel conductance	Misshaped CFTR pore restricts Cl <sup>-</sup> movement
V	Reduced CFTR protein production	Very low levels of CFTR protein
VI	CFTR protein turnover at cell surface	Functional but unstable CFTR protein at cell surface

Note. Adapted from "Targeted Therapies to Improve CFRT Function in Cystic fibrosis" by M. Brodlie, I. J. Hag, K. Roberts and J. S. Elborn, 2015, *Genome Medicine*, p. 2.

## Understand Cystic Fibrosis signs, symptoms and complications

"The signs and symptoms of cystic fibrosis (CF) vary from person to person (-)" (NHLBI, 2018).

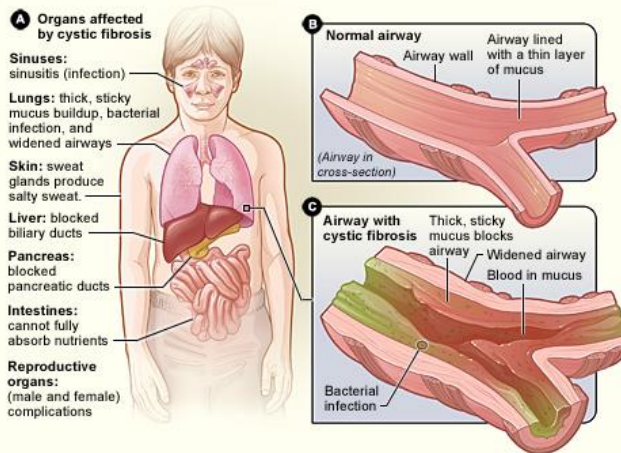
### Respiratory symptoms

- ✓ Persistent cough that produces thick mucus
- ✓ Wheezing
- ✓ Breathlessness
- ✓ Exercise intolerance
- ✓ Repeated lung infections
- ✓ Inflamed nasal passage or stuffy nose (Mayo clinic, 2016)

Symptoms vary over time and as disease gets worse, they are more severe (NHLBI, 2018).

### Digestive symptoms

- ✓ Foul-smelling greasy stools
- ✓ Poor weight gain and growth
- ✓ Intestinal blockage, particularly in newborns
- ✓ Severe constipation
- ✓ Rectal prolapse (Mayo Clinic, 2016)



## Diagnosis

In addition to clinical symptoms, doctors diagnose CF based on multiple tests including:

- ✓ Newborn screening (blood sample is checked for higher than normal levels of a chemical (immunoreactive trypsinogen, or IRT) released by the pancreas)
- ✓ Sweat test
- ✓ Other tests:
  - Genetic tests ( for type of CFTR defect)
  - Chest x-ray
  - Sinus x-ray
  - Lung functions test
  - Sputum culture
- ✓ Prenatal screening
- ✓ CF carrier testing ( detect faulty genes 9 out of 10 cases) ( NHLBI, 2018, diagnosis).

## Management

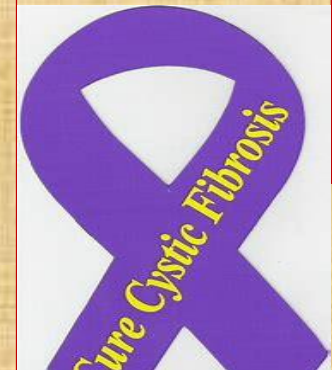
There is no cure. The goal of treatment include preventing and controlling lungs infection, loosening and removing thick an sticky mucus from lungs, preventing or treating blockage in the intestines, providing enough nutrition an preventing dehydration. This requires a multidisciplinary approach that involves a medical team of nurses, physical therapy , dietitians and social workers. Also, depending on the severity of symptoms, the CF treatment may requires long stay at he hospital (NHLBI, 2018, Treatment).

### Type of treatments

- ✓ Chest physical therapy that is called clapping or percussion involving pounding chest and back over and over with hands or devices
- ✓ Aerobic Exercise
- ✓ Treatment for advanced lung disease ( oxygen therapy or transplant)
- ✓ Pulmonary rehabilitation that include exercise training , nutritional counseling, education on lung disease or condition and how to manage it, energy-conserving techniques, breathing strategies, psychological counseling and/or group support
- ✓ Nutritional therapy with a well-balanced diet that's rich in calories, fat, and protein ( NHLBI, 2018)
- ✓ Traditional medicines:
  - Antimicrobial agents
  - Anti-inflammatory agents
  - Bronchodilators
  - Oral pancreatic enzymes (Mayo clinic, 2016, Medications).
- ✓ Precision medicines such as gene therapies for improving Transmembrane Regulator (CFRT) Function (e.g. Ivacaftor) (Murphy & Caraher, 2016, p.2) or gene therapy for restoration of normal CFTR function (Murphy & Caraher, 2016, p.9)
- ✓ Treatment of common complications such as diabetes and Osteoporosis (NHLBI, 2018).

## Conclusion

Cystic fibrosis is a complex disease that requires the dedication of healthcare providers that focus on providing specialized care and disease management. CF patients live depend on combination therapies that help loosen and get rid of the thick mucus that can build up in the lungs, open the airways or thin the mucus through inhaled medicines, and improve the absorption of vital nutrients by providing pancreatic enzymes supplement capsules (Cystic Fibrosis Foundation, 2018, treatment). However, as Garner, Gray, self and Wagener (2017) outlined, it is vital for patients to fully commit to their treatment to ensure positive outcomes by creating specialized individual plans. Care team must utilize strategies that increase communication with patients to "establish and document realistic, meaningful, and achievable goals; and ensure follow-up on progress and barriers to progress" (p.766). Finally, despite tremendous achievements in the fight for the cure of CF, too many lives are still being lost. And the more one understands the underlying mechanism of CF, the better equipped one is to handle this illness. In this perspective, genetic research with precision medicine offers the key to not only finding treatments that improve quality of life of CF patients but also in the fight of finding the cure for this terrible disease.



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