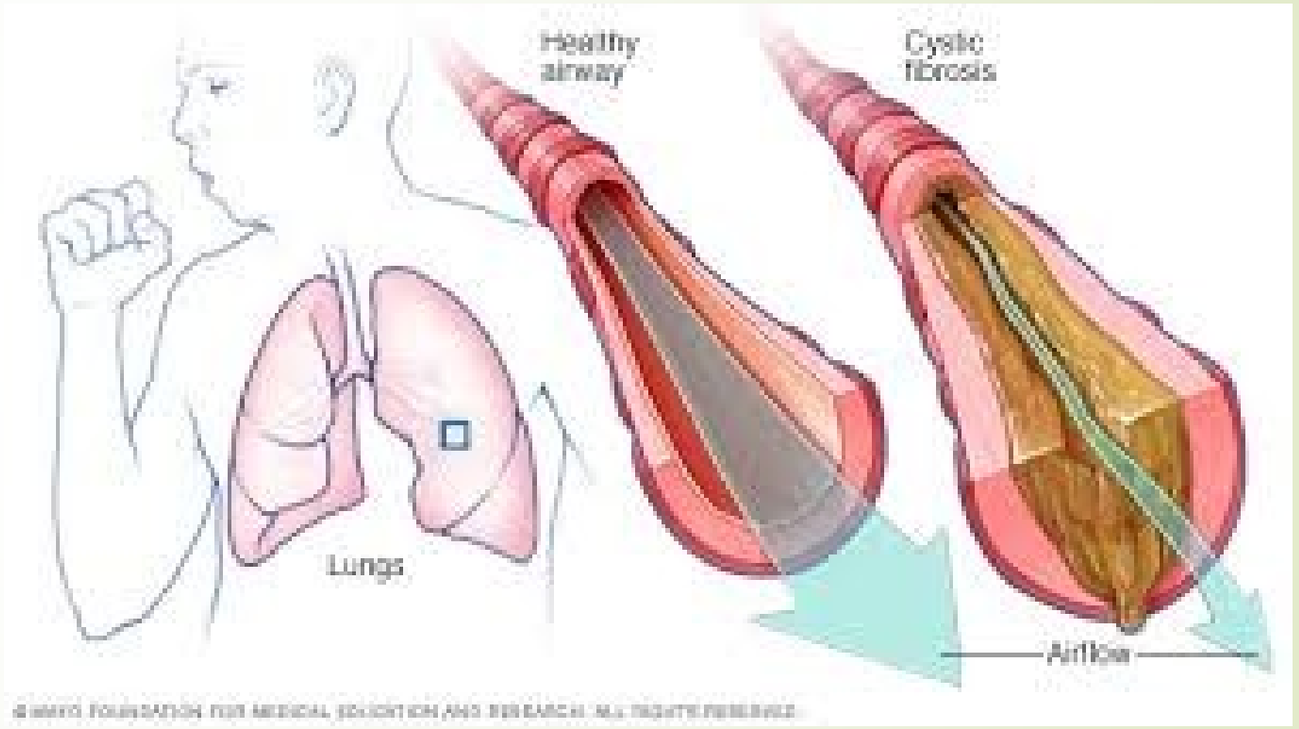
Cystic Fibrosis

Daniela Strasser

Pubudu Wanni Arachchige

What is it?

- Cystic Fibrosis is a heredity disease that affects the lungs, the digestive tract, pancreas, liver and kidneys
- The body produces thick and sticky mucus and secretions that can clog the lungs and obstruct the pancreas
- Different people have different degrees of symptoms
- There is no known cure



Etiology

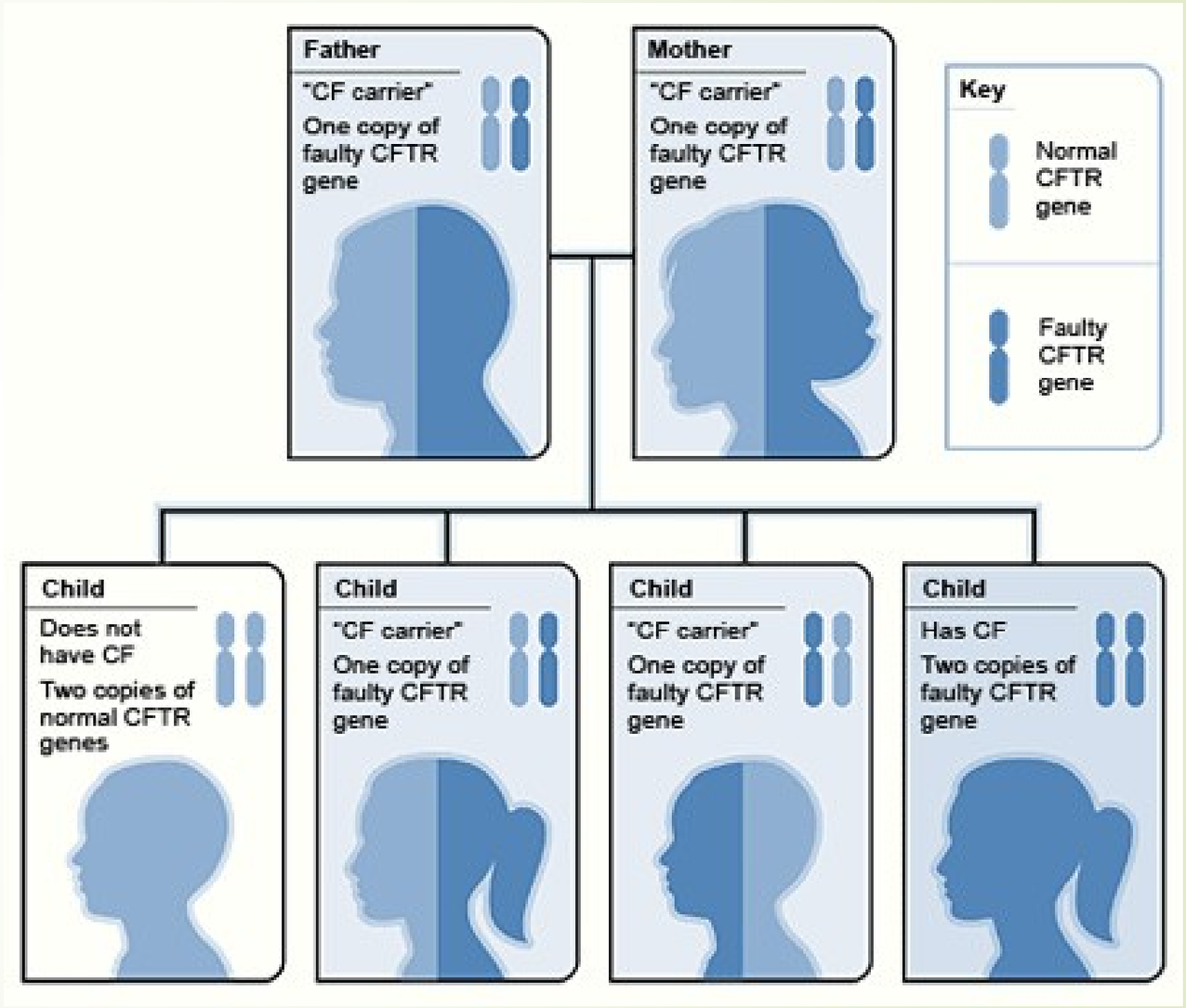
- CF is due to a mutation in the CF gene on chromosome 7
- The CF gene encodes a protein known as the cystic fibrosis transmembrane regulator (CFTR), which is a chloride channel essential in production of sweat, digestive juices and mucus.
- The abnormal CFTR protein in the patients with the disease leads to disruption of this chloride channels on the cells

Etiology

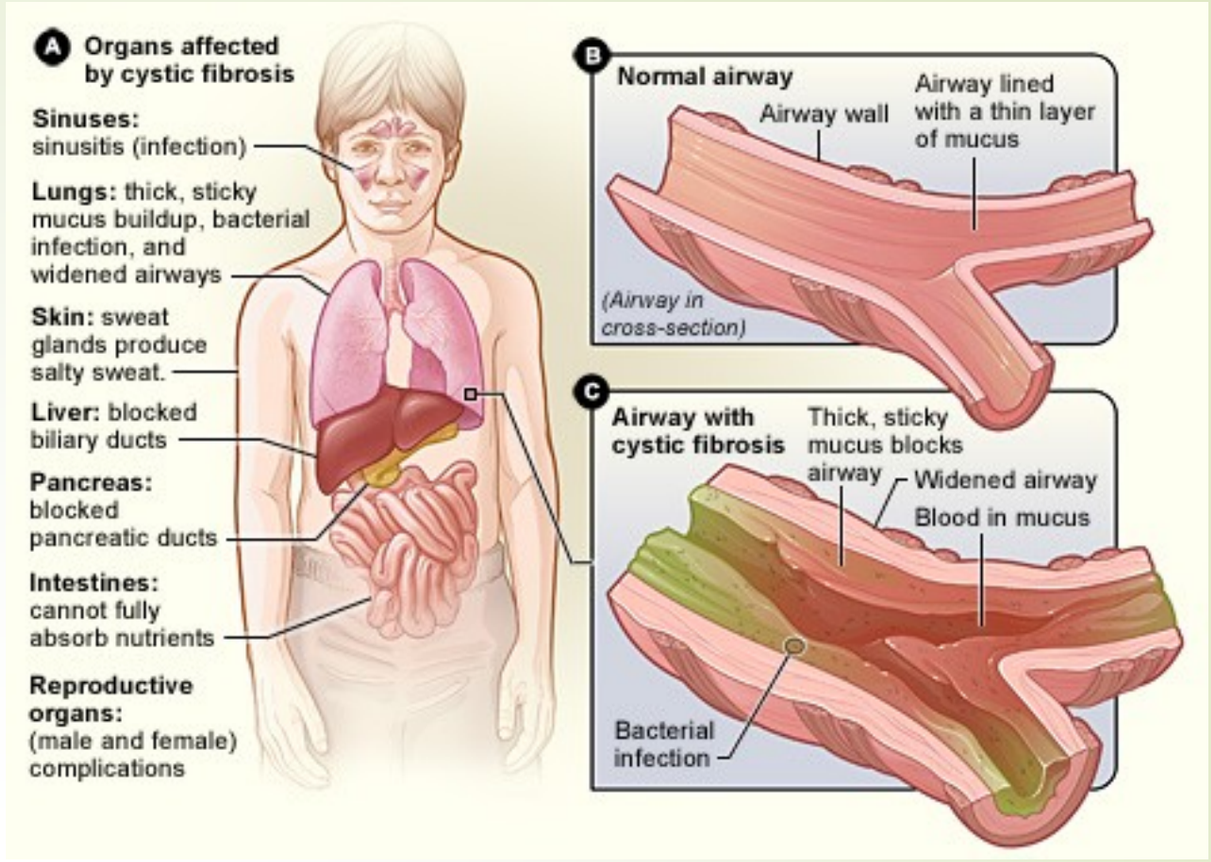
- Chloride channel disruption prevents them from regulating the flow of chloride ions and water across the cell membranes
- As a result, cells that line passageways of lungs, pancreas, etc. produce mucus that is unusually thick and sticky, clogging the airways and various ducts
- Several mutations of the gene can occur, which determines the severity of the symptoms

Etiology

- The disease is inherited autosomally recessive
- It means that the parents each carry one copy of the mutated gene but they don't show signs and symptoms



Symptoms



- Salty-tasting skin
- Persistent coughing
- Shortness of breath and wheezing
- Malnutrition and poor growth due to obstruction of the pancreatic digestive enzymes, which cause pancreatitis
- CF's obstruction of the lungs and impaired mucociliary clearance increase the risk of lung infections (bronchitis and pneumonia)
- Increased risk of diabetes and osteoporosis
- Infertility (male – congenital absence of vas deferens)
- Meconium ileus in newborns

Epidemiology

- CF is the most common life-limiting autosomal recessive disease among people of European heritage
- around 1 in 25 people of Northern European descent and 1 in 30 of Caucasian Americans is a carrier of a CF mutation
- Although technically rare, CF is ranked as one of the most widespread life-shortening genetic diseases

Diagnosis

- Newborn screening using a genetic test or a blood test. The genetic test shows whether a newborn has faulty CFTR and the blood test shows newborn's pancreas function

- Sweat test: doctor triggers sweating by applying pilocarpine on skin and then he uses and electrode to provide a mild electrical current to deliver the medication through he skin. The sweat is collected on a pad/paper and then analyzed . High levels of salt confirm diagnosis of cystic fibrosis

- Couple planning for pregnancy or are expecting can test for CFTR mutations and calculate the risk for the child

Treatment

- There is definite cure but management of the disease can allow survival well into adulthood
- Antibiotics: many CF-patients are on one or more ATB at all times, even when healthy, to prophylactically suppress infection
- Airway clearance: for example postural draining and percussion to get rid of mucus in lungs, chest physiotherapy
- Aerosolised medication loosen secrections like hypertonic saline

Treatment

- Nutritional therapy: pancreatic enzyme supplements, vitamin supplements
- Ibuprofen and azithromycin have been found to preserve and improve lung function
- CFTR modulators: newer drugs targeting faulty CF-causing gene
- Lung transplant

Sources

- https://www.medicalnewstoday.com/articles/147960#treatment
- https://www.medicinenet.com/cystic_fibrosis/article.htm
- https://www.cff.org