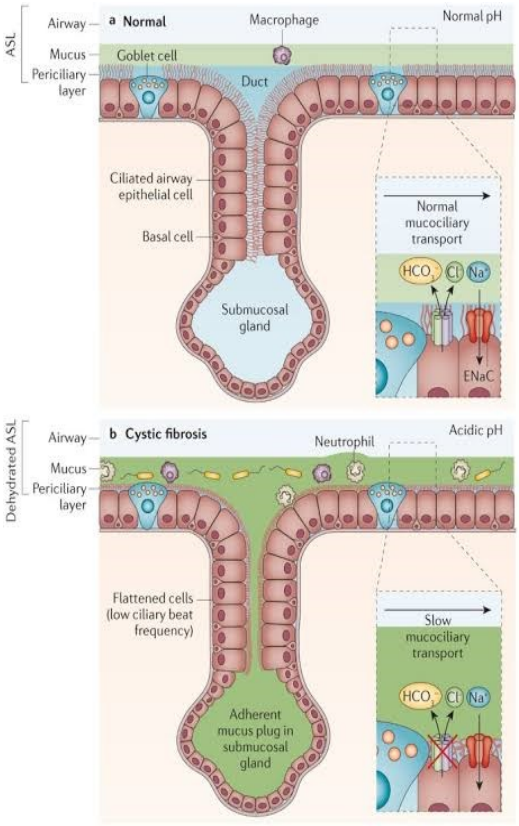
Cystic Fibrosis

Hiba Fatima Zaidi

1st year MBBS, Islamabad Medical and Dental College, Islamabad Pakistan

Key Points
➢ Common cause of chronic lung disease. ➢ Upper and lower respiratory tract pancreas and reproductive organs are affected.

Cystic Fibrosis is the most common cause of chronic lung disease in young adults. It is one of the most common fatal autosomal recessive diseases that affects about one in three thousand people.[1] This disease is distributed in variety of population and not only it affects Americans but also affects many south Asians and Africans.[2] It affects the upper and lower respiratory tract, pancreas and reproductive organs.



Source: *Nature reviews / Disease primers*

Patho-physiology:

It is caused by mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) protein due to which there is altered chloride transport and water flux across the epithelial surface of the exocrine glands this results in thickening of the secretions in multiple organs of the body. This results in abnormal mucus and that obstructs glands and ducts and lead to tissue damage.[3]

Sign & Symptoms:

The system manifestation ranges from growth retardation and chronic lung disease to chronic pancreatitis or infertility. In lungs cough, sputum production and hemoptysis are the most common symptoms. Patient may also suffer from steatorrhea, diarrhea and abdominal pain. The patients are malnourished and have low body mass index. On examination the findings may include clubbing, hyper resonant percussion notes and crackles. Most of the patients of cystic fibrosis has azoospermia due to the congenital bilateral absence of vas deferens. Along with these features biliary cirrhosis and gall stones may also be present. The infant and children suffering from CF may present with Meconium ileus, respiratory symptoms and failure to thrive in 20 %, 45 % and 28 % of the patient respectively.[4]

Diagnosis:

In order to diagnose CF, you need to have clinical findings along with biochemical or genetic conformation. The mainstay of diagnosis is sweat chloride test. However, test for specific mutation and

nasal potential difference. Diagnostic criteria for CF are as follow.[5]

- Clinical symptoms consistent with CF in at least one organ system, or positive newborn screen or having a sibling with CF
- Evidence of cystic fibrosis transmembrane conductance regulator (CFTR) dysfunction (any of the following):
- Elevated sweat chloride ≥60 mmol/L
- Presence of two disease-causing mutations in the CFTR gene, one from each parental allele
- Abnormal NPD

Molecular testing for CFTR gene mutation is used for conformation and it is also done in patients who have intermediate sweat chloride test results.

Pulmonary function test Chest X-ray and CT scan often required for the management of the patient.

Treatment:

As there are multiple systems involved in this disease so it requires a multi-disciplinary approach to care. Most of the time focus is on the pulmonary therapies but it is very important to pay attention to the nutritional status, Psycho social issues and glucose control of the patient.

Medications that are used to treat CF include the following:

- Pancreatic enzyme supplements
- Multivitamins (including fat-soluble vitamins
- Mucolytics
- Nebulized, inhaled, oral, or intravenous antibiotics
- Bronchodilators
- Anti-inflammatory agents
- Agents to treat associated conditions or complications e.g., insulin, bisphosphonates
- Agents devised to reverse abnormalities in chloride transport e.g., ivacaftor
- Inhaled hypertonic saline

The definitive treatment for advance CF is lung transplant.[6]

Patients should be immunized against pneumococcal and influenza infection. Family members of the affected individuals should be screened for CF and genetic counseling are also suggested.

Prognosis:

The median survival age of patient suffering from CF is now over 39 years and the patient usually die from pneumonia and cor pulmonale.

References:

1. FitzSimmons S. Cystic Fibrosis Foundation patient registry annual data report. Cystic Fibrosis Foundation Patient Registry Annual Data Report. 1997.
2. Sullivan BP, Freedman SD. Seminar cystic fibrosis. Lancet. 2009;373:1891-904..
3. Gibson RL, Burns JL, Ramsey BW. Pathophysiology and management of pulmonary infections in cystic fibrosis. American journal of respiratory and critical care medicine. 2003 Oct 15;168(8):918-51..
4. Accurso FJ, Sontag MK, Wagener JS. Complications associated with symptomatic diagnosis in infants with cystic fibrosis. The Journal of pediatrics. 2005 Sep 1;147(3):S37-41.
5. Farrell PM, White TB, Ren CL, Hempstead SE, Accurso F, Derichs N, Howenstine M, McColley SA, Rock M, Rosenfeld M, Sermet-Gaudelus I. Diagnosis of cystic fibrosis: consensus guidelines from the cystic fibrosis foundation. The Journal of pediatrics. 2017 Feb 1;181:S4-15.
6. Ramos KJ, Smith PJ, McKone EF, Pilewski JM, Lucy A, Hempstead SE, Tallarico E, Faro A, Rosenbluth DB, Gray AL, Dunitz JM. Lung transplant referral for individuals with cystic fibrosis: Cystic Fibrosis Foundation consensus guidelines. Journal of Cystic Fibrosis. 2019 May 1;18(3):321-33.