# CYSTIC FIBROSIS

## {Mucoviscidosis}

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## ➢ Just Remember These Important Notes ⬇⬇

- ➲ It is an inherited disease which is autosomal recessive (that skip generations)
- ➲ There are mutations affecting CFTR (CF transmembrane conductance regulator) gene on chromosome 7
- ➲ CFTR is responsible of transport of Chloride across the membranes of epithelial cells.
- ➲ In Cystic fibrosis, the chloride is not absorbed from the lumens of exocrine glands and surface epithelium.

  Likewise, Na absorption is also decreased, resulting in VISCID mucus secretions.

## ➢ Viscid Secretions In Different Organs ⬇⬇

### I. In lungs:

Recurrent chest infections & pneumonias ---> end in Bronchiectasis

Bronchiectasis was discussed before in details

### II. in GIT :

Distal intestinal obstruction syndrome (acute surgical abdominal pain + absolute constipation)

**In Neonates:** Meconium ileus syndrome

### III. In Pancreas :

Decrease in both exocrine & endocrine secretions of Pancreas

(Pancreatic insufficiency)↴

- **Exocrine:** Decreased lipase & amylase ↴↴
  - Steatorrhea & weight loss
  - Epigastric pain
  - Failure to thrive is common in children with Cystic fibrosis.
- **Endocrine** : Decreased Beta cells secretions ---> Low insulin: ---> **{Diabetes}**

### IV. Biliary tract & liver :

Concentrated bile & damage to biliary ducts ⇩
Stones formation ⇩
Obstructive jaundice ⇩
Biliary Cirrhosis

### V. Nasal sinuses : Sinusitis & polyps

### VI. Genital organs:

- Defect in Vas deference in males, leading to infertility
- The disease starts in neonatal period or infancy or childhood.
- The patients will be greatly manifested by the age of 15 - 20 (mostly chest problems, then Pancreas, then fertility)

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## ➢ DIAGNOSIS :

- Sweat chloride test (sweat is collected via pilocarpine iontophoresis)
  Level > 60 m.mol/L is diagnostic
- confirm diagnosis by Genetic test for Detection of mutation of ***CFTR*** gene

## ➢ Other important investigations ⇩

### ➲ For chest

- Do HRCT chest, spirometry
- ECHO to detect if there is pulmonary HTN or not as a complication of long standing bronchiectasis ( Cor pulmonale)

### ➲ For Pancreas

- Do fasting ,post prandial BG & HbA1C To detect if there is Diabetes
- Do fecal elastase ( for pancreatic insufficiency )
- Qualitative test for fat in stool ( Sudan III) quantitative test is difficult
- Basic labs& electrolytes for malabsorption.

### ➲ For biliary tract & liver

- Do US abdomen ± ERCP if obstructive jaundice is present.
- ALP ,GGT , bilirubin

➲ **For fertility:** Do semen analysis.

## ➢ TREATMENT:

- **Until now there is no curative treatment for it**
- **Gene therapy:** under trials
- **Treatment of organ damage**

**I. Chest:** Treatment of Bronchiectasis as mentioned before

⇨ Postural drainage, chest physiotherapy

⇨ Measures to decrease bacterial colonization & infective exacerbations

- Nebulized hypertonic saline,
- Nebulized Dornase alpha e.g. © ***Pulmozyme inh amp***

  It is recombinant DNAse acting by liquefaction of viscid mucus, thereby decreasing chances of repeated infections & improvement of respiratory functions.

⇨ there are certain bacteria, which are responsible for exacerbations ⇩

- Pseudomonas aeroginosa
- Burkholderia cepacia
- Staph aureus in children

So, we can use prophylactic antibiotics Such as Azithromycin

⇨ In cases of documented Pseudomonas infection ⇩

--> Secondary prevention by inhaled tobramycin

⇨ **Infective exacerbations**: as mentioned in bronchiectasis

**II. Pancreas :**

⇨ Treatment of DM by insulin

⇨ Treatment of pancreatic insufficiency by ⇩

- Pancreatic enzyme replacement
- Pancrelipase (CREON CAP ©)

**III. Hepatobiliary tract:**

ERCP if needed + stone removal & stents

**IV. GIT :** Distal intestinal obstruction Syndrome --> Surgical treatment

**V. Treatment of infertility.**