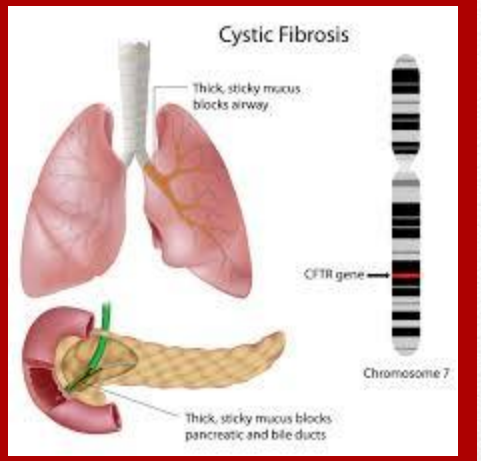

Cystic fibrosis

Clinical Presentation of Cystic Fibrosis

General

- Usually diagnosed in neonates (meconium ileus or newborn screening) or during early childhood. May present later in life due to less severe symptoms or misdiagnosis.

Symptoms

- Pulmonary: Chronic cough, sputum production, decreased exercise tolerance, and recurrent pneumonia and sinusitis. Exacerbations may be marked by increased cough, sputum changes (darker, thicker), hemoptysis, dyspnea, and fever.
- GI: Numerous large, foul-smelling loose stools (steatorrhea), flatulence, and abdominal pain. Intestinal obstruction may present as abdominal pain and distention and/or decreased bowel movements.
- Nutritional: Poor weight gain despite voracious appetite and hunger. Dry skin, skin rash, and visual disturbances may be noted in vitamin deficiency.
- CFRD: Weight loss, increased thirst, and more frequent urination.

Signs

- Obstructive airways disease: Tachypnea, dyspnea, cyanosis, wheezes, crackles, sternal retractions, digital clubbing, and barrel chest.
- Failure to thrive: Below age-based normal in both height and weight in children; adults may be near/below ideal body weight or have a low body mass index (BMI).
- Salty taste to the skin.
- Hepatobiliary disease: Hepatomegaly, splenomegaly, and prolonged bleeding may occur.
- Recurrent pancreatitis (usually in pancreatic-sufficient patients): Episodic epigastric abdominal pain, persistent vomiting, and fever.

Laboratory Tests

- Leukocytosis with increase in polymorphonuclear (PMN) leukocytes and bands may occur in acute pulmonary exacerbations.

- Maldigestion: Decreased serum levels of fat-soluble vitamins (A, D, E, and K). Decreased vitamin K levels may result in elevated prothrombin time (PT) and international normalized ratio (INR).
- Glucose intolerance: Blood glucose between 140 and 199 mg/dL (7.8–11.0 mmol/L) 2 hours after an oral glucose-tolerance test.
- CFRD: Blood glucose 200 mg/dL (11.1 mmol/L) or higher 2 hours after an oral glucose-tolerance test or fasting hyperglycemia (fasting blood glucose 126 mg/dL [7.0 mmol/L] or more regardless of the postglucose challenge level).
- Hepatobiliary disease: Serum aspartate aminotransferase, alanine aminotransferase, alkaline phosphatase, γ-glutamyltransferase, and bilirubin may be elevated.

Other Tests

- Microbial cultures (sputum, throat, bronchoalveolar lavage, or sinus): Isolation of *P. aeruginosa, S. aureus, S. maltophilia,* and other CF-related organisms.
- Pulmonary function tests (PFTs): Decreased forced expiratory volume in 1 second (FEV_1) and forced vital capacity (FVC), typically lower during acute pulmonary exacerbations.
- Chest x-ray or CT scan: Infiltrates, atelectasis, bronchiectasis, and mucus plugging.
- Abdominal x-ray or CT scan: Intestinal obstruction may be manifested as meconium ileus, DIOS, or intussusception. Rectal prolapse may be noted on physical examination.
- Maldigestion: Elevated fecal fat content, reduced pancreatic stool elastase (less than 200 mcg/g of feces).

Diagnosis

- Testing for CF is part of required newborn screening (immunoreactive trypsinogen-IRT) panels in all US states

All “positive screens,” as well as individuals presenting with signs and symptoms of CF, are referred to a CF care center for **sweat chloride test and genetic evaluation**.

Pilocarpine iontophoresis (“sweat test”)