Cystic Fibrosis

CF is an **autosomal recessive** genetic condition characterized by mutations of the **cystic fibrosis transmembrane conductance regulator (CFTR) gene** found on chromosome 7. The most common mutation is **delta F508 deletion**.

PATHOPHYSIOLOGY

Dysfunction of the CFTR protein found in epithelial cells of respiratory, gastrointestinal, reproductive system & sweat glands ⇢ dysregulation of chloride, bicarbonate, & sodium channel transport ⇢ ↓ H2O concentration of secretions ⇢ impaired clearance

EPIDEMIOLOGY

Most prevalent in Caucasians ⇢ ~ 1 in every 3000 – 3500 live births

CLINICAL MANIFESTATIONS

Respiratory

- Progressive disease with intermittent acute exacerbations
- Mucous plugging ⇢ chronic airway inflammation & infections ⇢ bronchiectasis
- **Common pathogens:** S. aureus, H. influenza, P. aeruginosa, B. cepacia

Gastrointestinal

- Intestinal obstruction
- Steatorrhea
- Pancreatic insufficiency, initially exocrine ⇢ eventually also endocrine (CF related diabetes)
- Fat soluble vitamin deficiencies (ADEK)
- CF related liver disease

Other

- Failure to thrive
- Delayed puberty & infertility
- Dehydration & sodium depletion
- Stress incontinence
- Low bone mineral density
- Clubbing

INVESTIGATIONS

- Newborn screening test
- Sweat chloride test
- CF genetic testing

MANAGEMENT

- Airway clearance techniques: chest physiotherapy, nebulized hypertonic saline, dornase alfa
- Regular pulmonary function tests
- Lung transplantation
- Nutrition to maintain BMI >50th percentile
- Pancreatic enzyme supplementation
- Fat soluble vitamin supplementation (ADEK)
- Increased salt intake
- Oral glucose tolerance tests
- CFTR modulator therapies
- Routine immunizations
- Exposure avoidance (no contact with other individuals with CF; no hot tubs or humidifiers)
- Mental health screening (patient & family)

Published February 2021
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