
CYSTIC
FIBROSIS
A Brief Overview

- **Autosomal Recessive disorder**
- Mutations of a single gene on the long arm of chromosome 7
- Defect in Cystic fibrosis trans-membrane regulator (CFTR)
- The major effects are on the lungs and pancreatic exocrine function
- Combination of defective chloride secretion and increased sodium absorption across airway epithelium
- Predispose the lung to chronic pulmonary infections and bronchiectasis

- 1:2000 live births
- Affecting Caucasians.
- 1:25 people carry a copy of the faulty gene.

Clinical Features

- **Neonate:** Failure to thrive; meconium ileus; rectal prolapse.

Clinical Features

- **Children and young adults:**
  - **Respiratory:** cough; wheeze; recurrent infections; bronchiectasis; pneumothorax; haemoptysis; respiratory failure; cor pulmonale
  - **Gastrointestinal:** pancreatic insufficiency (diabetes mellitus, steatorrhoea); distal intestinal obstruction syndrome (meconium ileus equivalent); gallstones; cirrhosis
  - **Others:** male infertility; osteoporosis; arthritis; vasculitis; nasal polyps; sinusitis; and hypertrophic pulmonary osteoarthropathy (HPOA)

Clinical Features

- **Signs**
  - Cyanosis
  - Finger clubbing
  - Bilateral coarse crackles.

Diagnosis

- **Sweat test:** Sweat sodium and chloride >60mmol/L
- **Genetics:** Screening
- Faecal elastase: Screening test for exocrine pancreatic dysfunction

Other Tests

- FBC
- U&E
- LFT
- Clotting
- Vitamin A, D, E levels
- Annual glucose tolerance test

Other Tests

- *Bacteriology*: Cough swab, sputum culture
- *Radiology:*
  - CXR: hyper inflation; bronchiectasis
  - Abdominal ultrasound: Fatty liver; cirrhosis; chronic pancreatitis
- Spirometry: Obstructive defect
- Aspergillus serology/skin test
- Faecal fat analysis

Management

- **Multidisciplinary approach**
- **Chest:**
  - Physiotherapy (postural drainage, airway clearance techniques)
  - **Antibiotics** are given for acute infective exacerbations and prophylactically
  - Chronic Pseudomonas infection is an important predictor of survival
  - **Mucolytics** - DNase, or nebulized hypertonic saline
  - **Bronchodilators**
  - *Advanced lung disease:* Oxygen, diuretics (cor pulmonale); noninvasive ventilation; lung or heart/lung transplantation

Management

- **Gastrointestinal:**
  - Malabsorption, GORD, distal obstruction syndrome
  - **Pancreatic enzyme** replacement
  - **Fat-soluble** vitamin supplements (A, D, E, K)
  - **Ursodeoxycholic acid** for impaired liver function; cirrhosis may require liver transplantation.

Management

- **Other:**
  - Treatment of CF-related **diabetes** (screen annually with OGTT from 12yrs)
  - Screening/treatment of **osteoporosis**
  - Management of **Arthritis**, **sinusitis**, and **vasculitis**
  - **Fertility**
  - **Genetic counselling**

Management

- **Ivacaftor** and **Lumacaftor** target the CFTR protein
- Ivacaftor, a CFTR potentiator
- Lumacaftor is a CFTR corrector

Prognosis

- Median survival is now ~ 41yrs in the UK
- Although a baby born today would expect to live longer

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