

Cystic Fibrosis

A

- Autosomal recessive condition. 1 in 25 are carriers (in UK) 1 in 2500 affected.
- Abnormal genes on Chromosome 7 which produce CFTR protein, most commonly defect is ΔF508

S+S

- Failure to thrive
- Malabsorption
- Recurrent Chest infections
- Harrison's Sulci
- Infertility in males
- Pneumothorax (es)
- Diabetes
- Mæconium ileus
- Bronchiectasis
- Hyperinflated Chest
- Finger Clubbing
- Steatorrhea
- Depression
- School Absences
- Hæmoptysis
- Productive cough
- Cholesterol Gallstones
- Pneumonia
- 'Salty Taste'
- Nasal Polyps

P

- CFTR = Cystic Fibrosis transmembrane regulator (Na/Cl pump)
- Its absence gives ↑viscosity of secretions
- Also effects on inflammatory processes and the immune response.
- infertility in males is due to absent vas deferens

I

- Sweat test → sweating induced by pilocarpine iontophoresis, sweat collected, +ve if NaCl concentration 60+, high false +ve and false -ve rates
- Guthrie test as neonate. •Bloods (FBC, LFT, U+E, Malabsorption screen, glucose)
- Genetic screening: •CXR
- Sputum culture

T

- ↑life expectancy!
- MDT!!!
- Annual review in tertiary centres
- Physiotherapy BD
- Abx (?Prophylaxis, may need central venous catheter).
- Regular nebs (saline)
- Counselling
- Gene therapy (very new)
- Diet = ↑calorie ↑protein ↓fat
- Creon (pancreatic enzymes) with meals
- Vitamin supplements
- Lung/Liver transplantation

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