TOPIC: Cystic Fibrosis

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Definition & Epidemiology:

Cystic fibrosis (CF) is a lethal autosomal recessive disorder affecting ion transport in exocrine glands, leading to thick, obstructive secretions. It is most common in Caucasians (1 in 2,500 live births).

Cause:

Mutations in the CFTR gene (chromosome 7q31.2) impair chloride and bicarbonate transport, particularly affecting lungs, pancreas, GI, and reproductive tracts.

Pathophysiology:

CFTR regulates ion channels including ENaC. Mutation leads to increased sodium/water absorption → dehydrated mucus → obstruction and infections. Sweat glands show decreased NaCl reabsorption → salty sweat. Impaired bicarbonate secretion results in acidic secretions, mucus plugging, and bacterial adhesion.

Genetics:

CFTR mutations are classified into six classes:

I–III: Severe (no/defective protein)

IV–VI: Mild (residual function)

Clinical Features:

Respiratory: Chronic infections (esp. Pseudomonas, Burkholderia), bronchiectasis, ABPA

Gastrointestinal: Pancreatic insufficiency, steatorrhea, meconium ileus

Hepatic: Steatosis, focal biliary cirrhosis

Reproductive: Male infertility due to CBAVD

Diagnosis:

Elevated sweat chloride (>60 mM)

Newborn screening (↑ immunoreactive trypsinogen)

Genetic testing (CFTR sequencing)

Nasal potential difference test

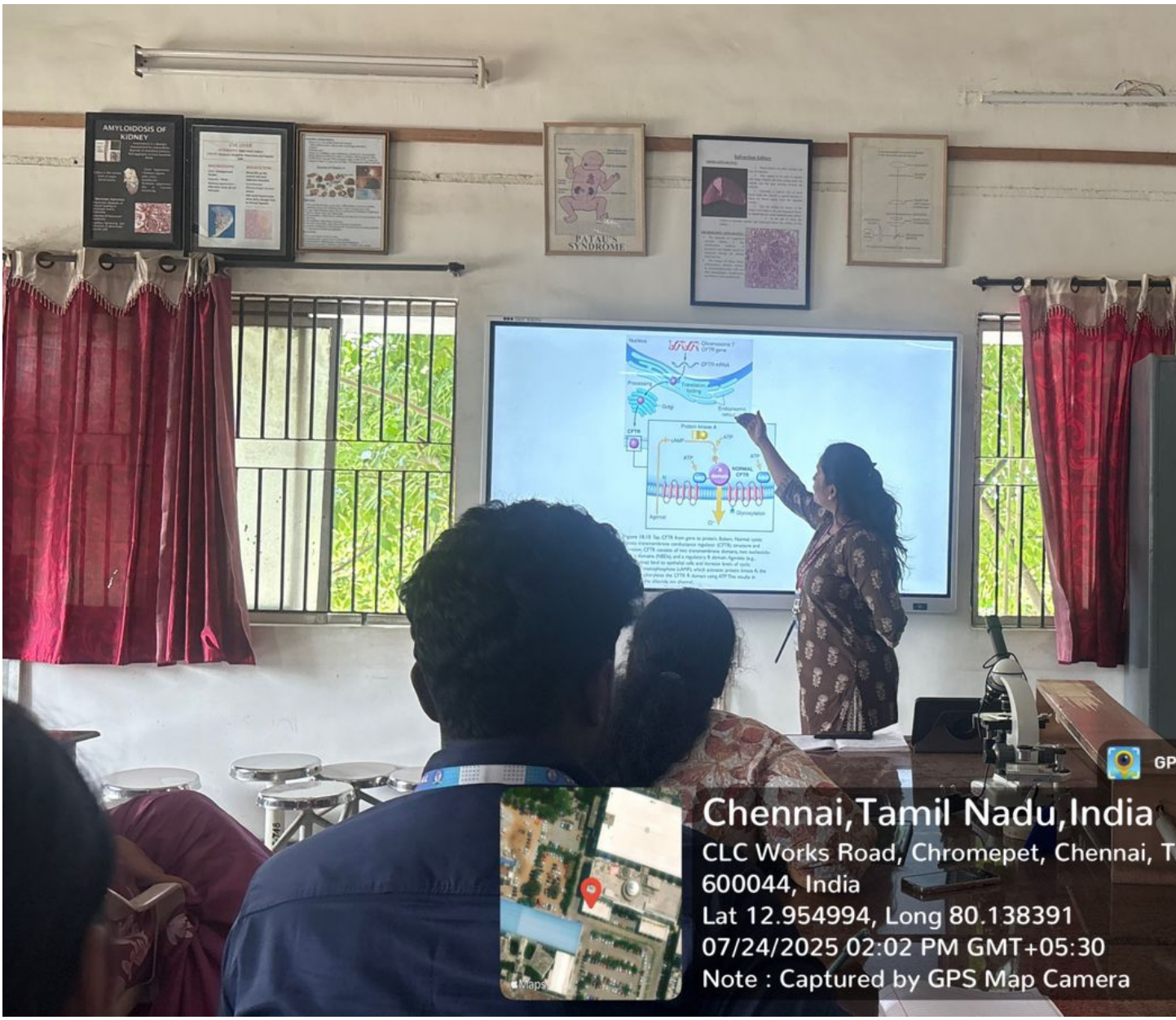
Treatment:

Antibiotics for infections

Pancreatic enzyme supplements

Lung transplant (in severe cases)

CFTR modulators (based on mutation class)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