Cystic Fibrosis (CF)

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Course: Genetics II (MA 206)

Summer Class

Lecture 7

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Outline

- What is Cystic Fibrosis
- Mechanism of Disease
- Common CFTR Mutation
- Treatment and Management

Objectives

- By the end of this lecture, students should be able to:
- Explain the genetic basis of cystic fibrosis (CF)
- Describe the molecular mechanism of CFTR protein dysfunction.
- Link CFTR mutations to clinical symptoms.
- Discuss current treatment strategies targeting CF mechanisms.

❑ What is Cystic Fibrosis?

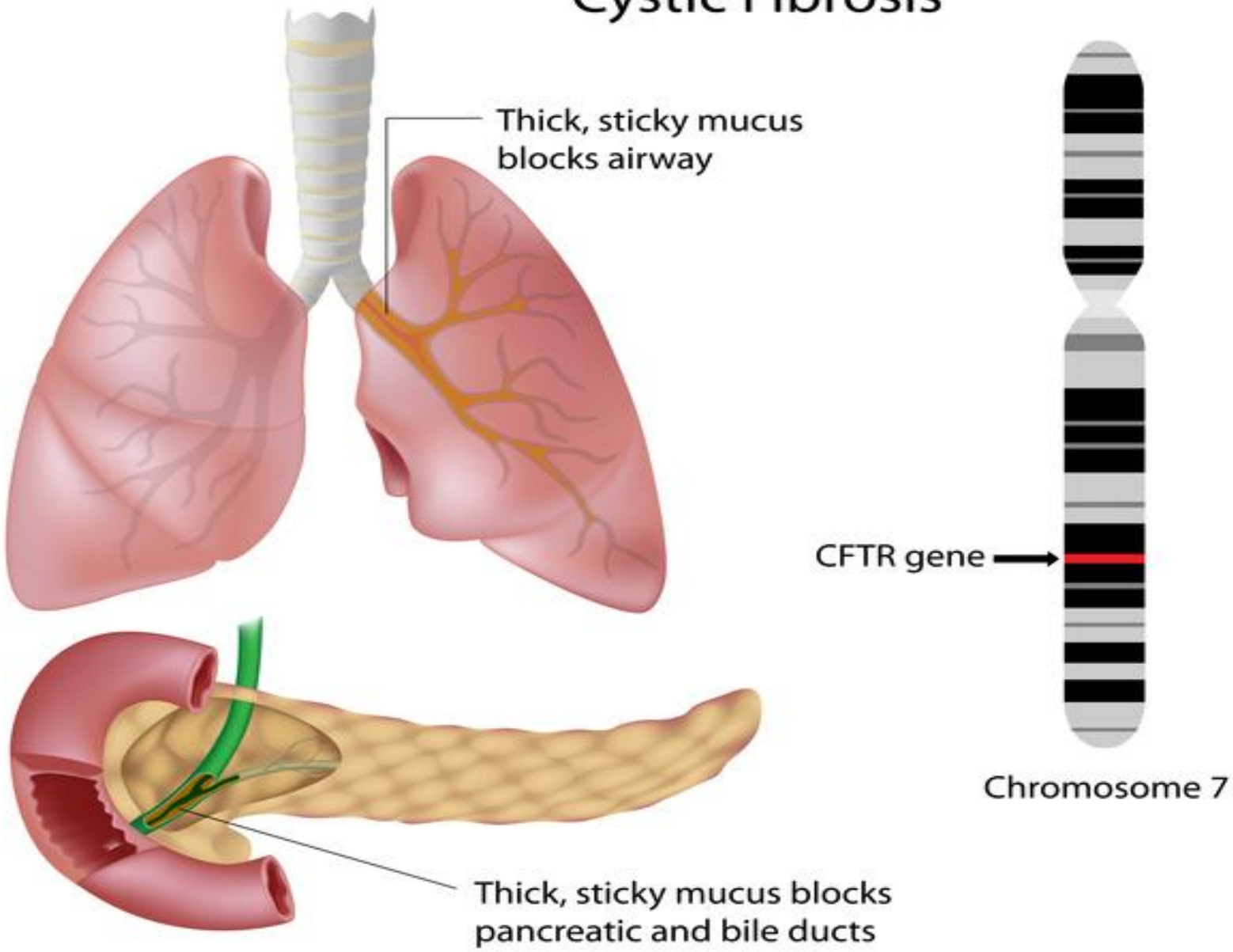
- Is a genetic disorder that primarily affects the **respiratory**, **digestive**, and **reproductive systems**.

- It is caused by mutations in the **CFTR gene**, causing thick, sticky mucus to build up in various organs.

- Affected organs: Lungs, pancreas, intestines, liver, reproductive tract, and sweat glands.

- CF is one of the most common **life-threatening inherited** diseases.

Cystic Fibrosis



❖ CFTR Gene and Protein:

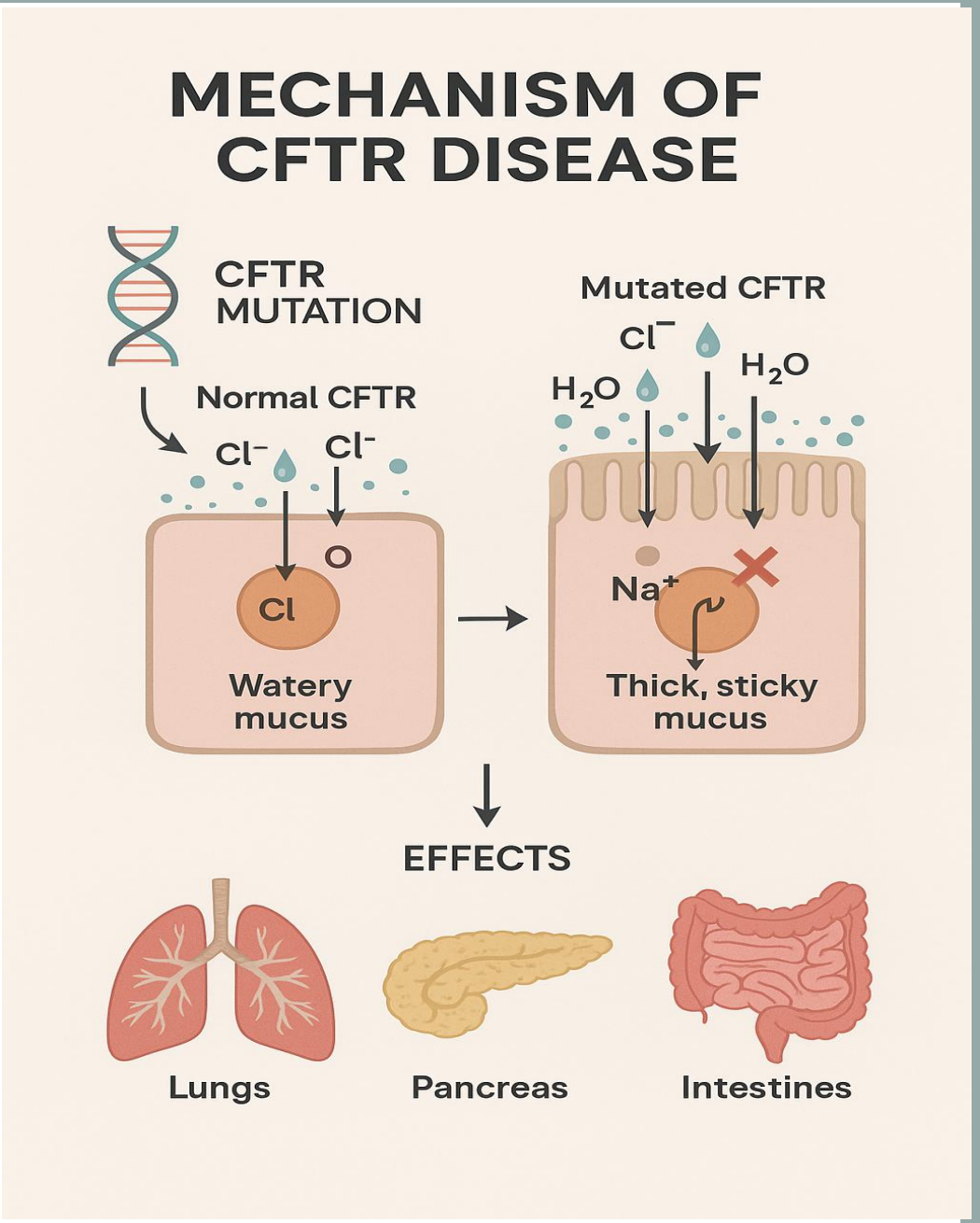
- CFTR stands for **Cystic Fibrosis Transmembrane Conductance Regulator**.

i. Located on **chromosome 7 (7q31.2)**.

ii. Encodes a **chloride channel protein**.

iii. Regulates the movement of **chloride and sodium ions**.

- Mutated CFTR: Chloride channels are defective or absent.
- Chloride and water movement is reduced.
- Mucus becomes thick, sticky, and difficult to clear.
- Leads to blockages, inflammation, and infections.



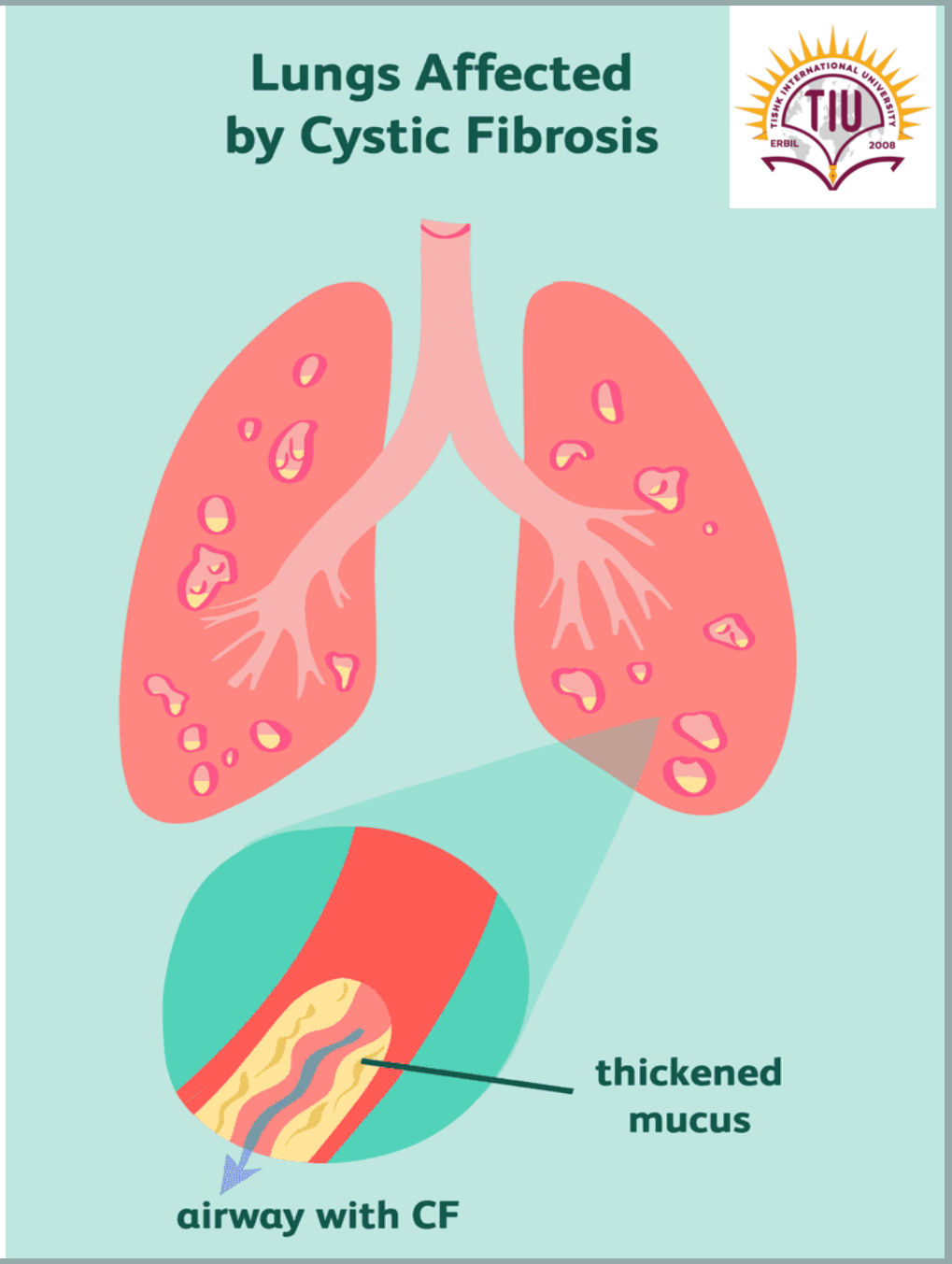
❑ Common CFTR Mutation:

❖ The most common mutation: **ΔF508** (Deletion of phenylalanine at position 508).

❖ Affects protein folding and trafficking, causing it to degrade before reaching the membrane.

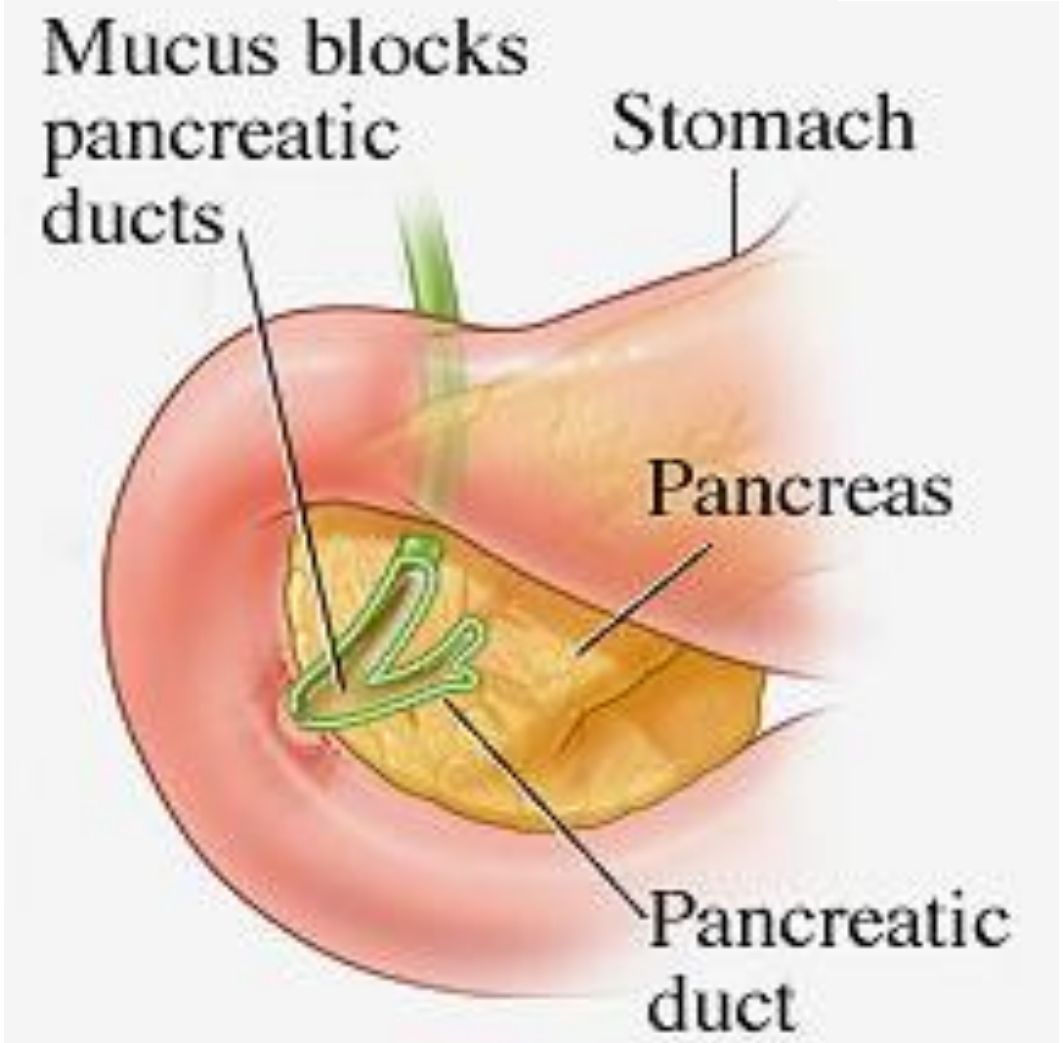
❑ Respiratory System Effects:

- Thickened mucus in airways leads to chronic lung infections, bronchiectasis, and reduced lung function.
- Increased susceptibility to bacterial infections.
- Progressive respiratory failure.



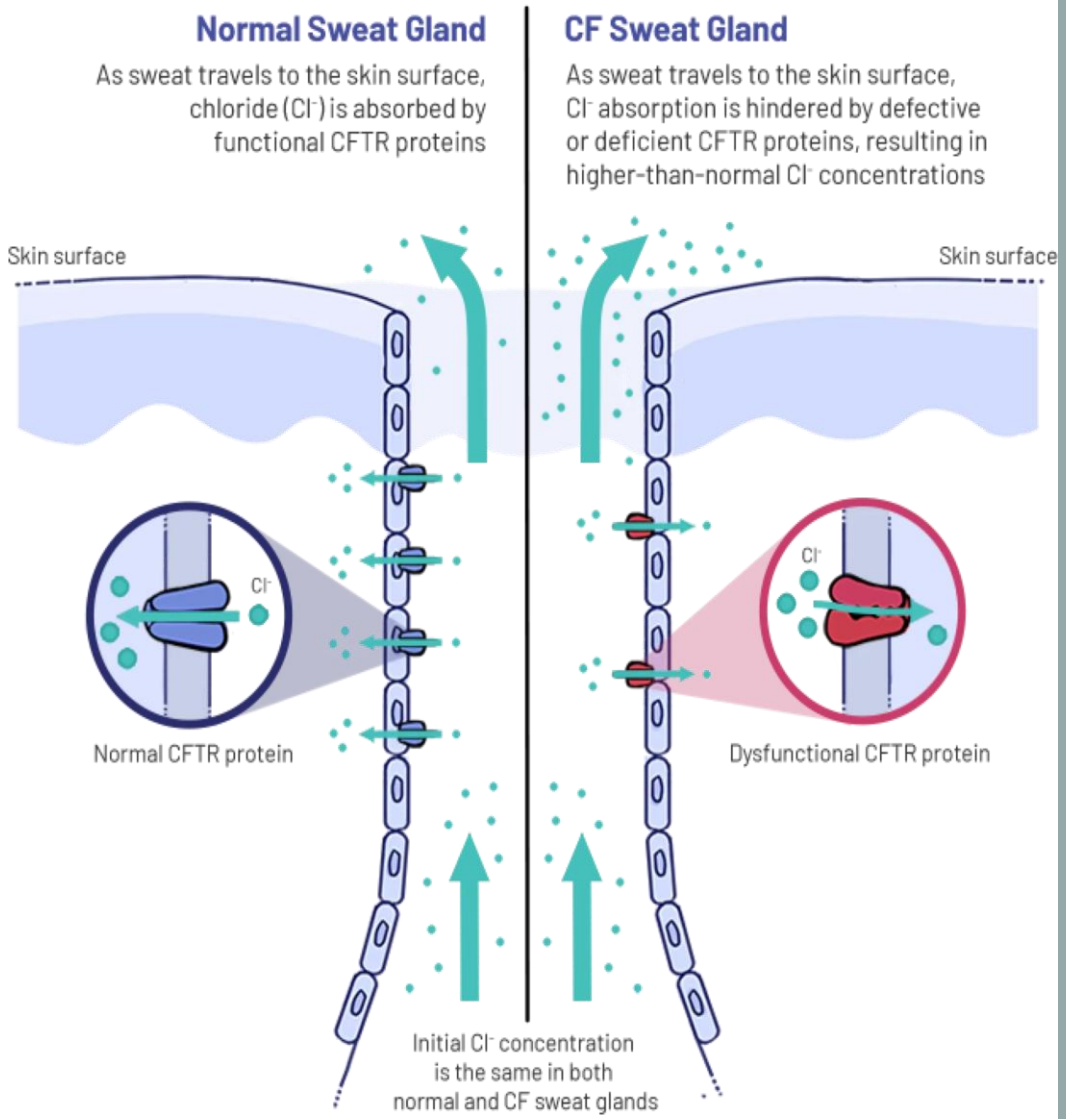
❑ Digestive System Effects:

- Pancreatic insufficiency.
- Meconium ileus in newborns.
- Increased risk of diabetes mellitus.



❑ Sweat Gland Dysfunction:

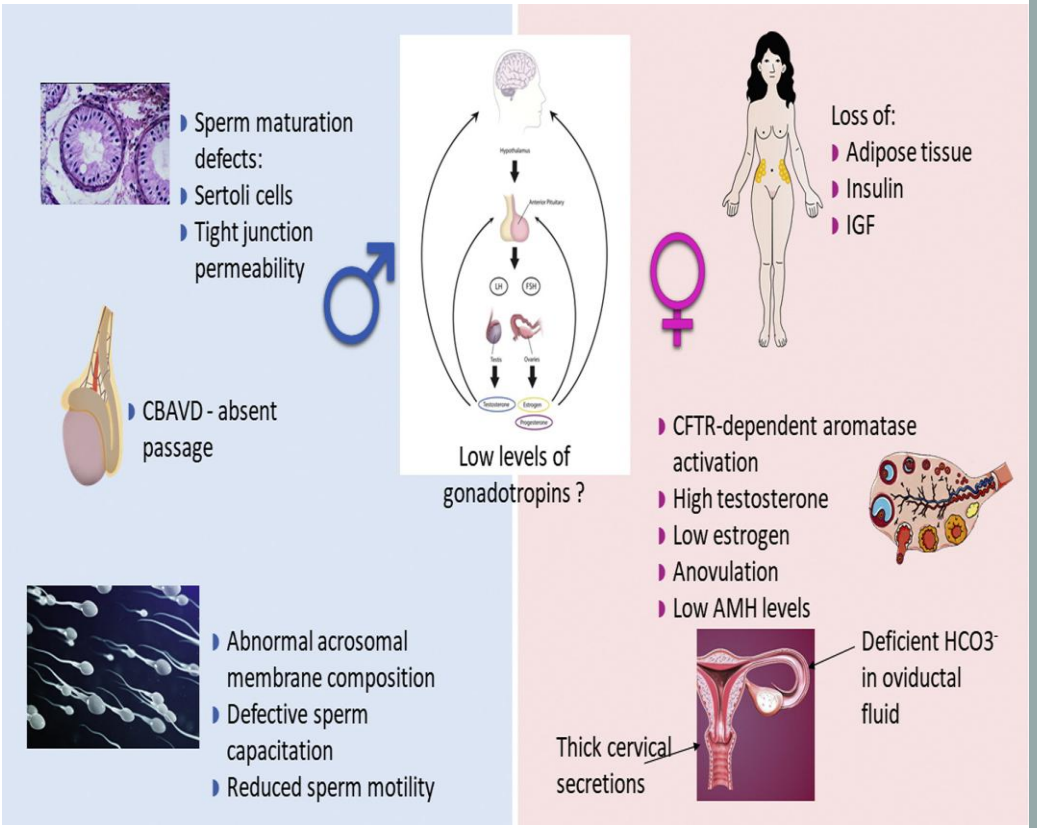
- Excessive chloride and sodium loss in sweat leads to salty skin, dehydration, and electrolyte imbalances.



❑ Reproductive System Effects:

- Males: Congenital bilateral absence of the **vas deferens** (CBAVD) leads to infertility.

- Females: Thick cervical mucus may cause reduced fertility.



❑ Organs Affected and Symptoms:

System	Major Effects	Common Symptoms
Respiratory	Mucus buildup, bacterial infection	Chronic cough, wheezing, lung infections
Digestive	Blocked pancreatic ducts	Poor weight gain, malabsorption, steatorrhea
Reproductive	Blocked vas deferens in males	Infertility
Sweat glands	Elevated salt in sweat	Salty skin, used in diagnostic testing

❑ Diagnosis of Cystic Fibrosis:

1. Newborn Screening

2. Sweat Chloride Test

3. Genetic Testing

4. Lung Function Tests

❑ Treatment and Management:

- Airway Clearance Therapy
- Antibiotic Therapy
- Pancreatic Enzyme Replacement Therapy (PERT)
- CFTR Modulators
- Nutritional Support
- Lung Transplant

References

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3. Vafai SB, Mootha VK. "Mitochondrial disorders as windows into an ancient organelle." Nature, 2012.

Thanks