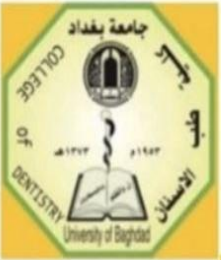


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# Cystic Fibrosis Disease Causes and Symptoms

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- Cystic fibrosis (CF) is a genetic condition that affects a protein in the body. People who have cystic fibrosis have a faulty protein that affects the body's cells, its tissues, and the glands that make mucus and sweat. Normal mucus is slippery and protects the airways, digestive tract, and other organs and tissues.
  - Cystic fibrosis causes mucus to become thick and sticky. As mucus builds up, it can cause blockages, damage, or infections in affected organs.

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- Cystic fibrosis used to cause death in childhood. Survival has improved because of medical discoveries and advances in newborn screening, medicines, nutrition, and lung transplants. Nearly 40,000 children and adults in the United States and more than 100,000 worldwide are now living with cystic fibrosis. Children born between 2018 and 2022 who have cystic fibrosis are expected to live an average of 56 years. On average, half of babies born in 2021 with cystic fibrosis are expected to reach the age of 65 or older.

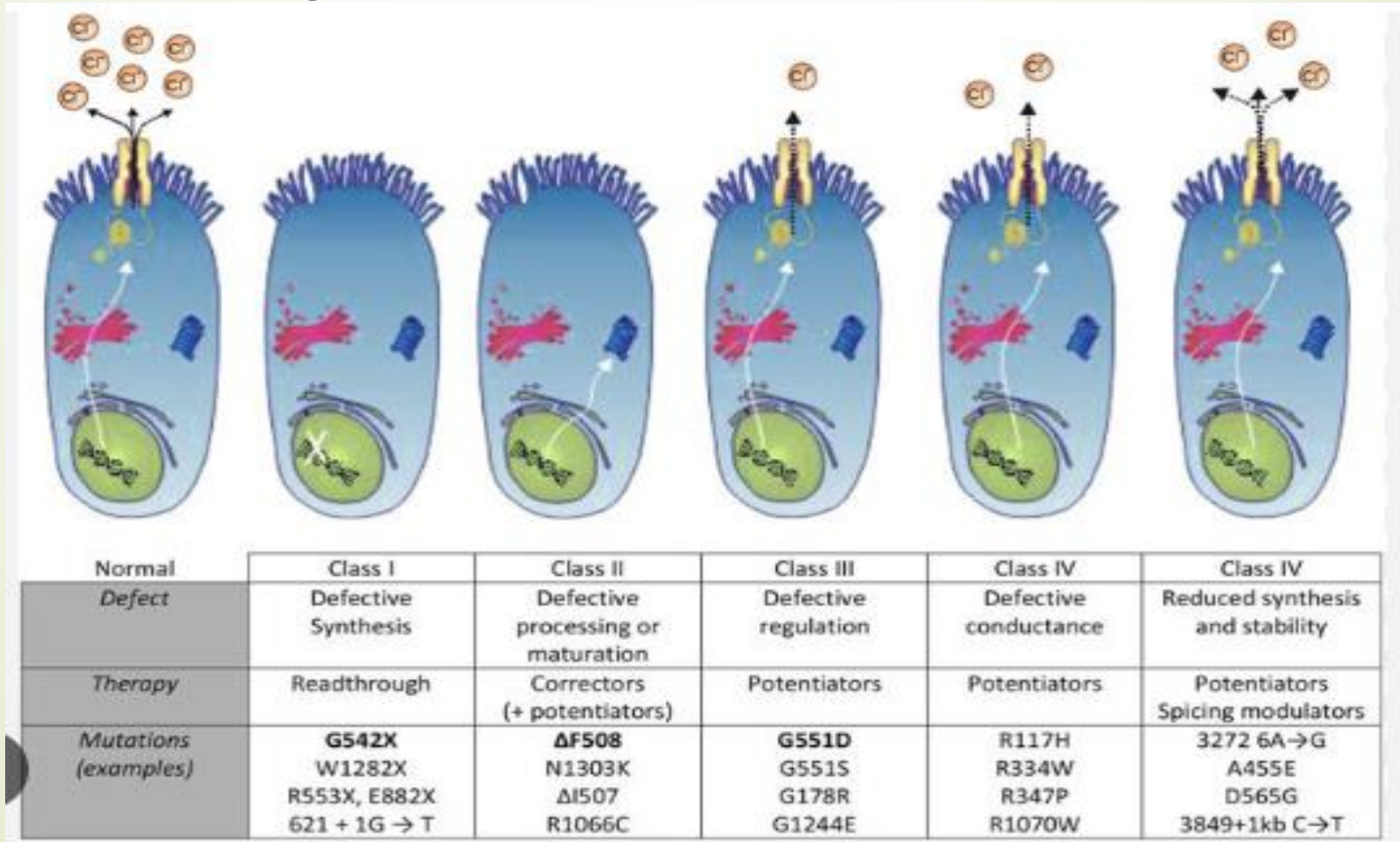
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- ▶ People with cystic fibrosis have a higher than normal level of salt in their sweat. Parents often can taste the salt when they kiss their children. Most of the other signs and symptoms of CF affect the respiratory system and digestive system.
  - ▶ Children need to inherit one copy of the gene from each parent in order to have the disease. If children inherit only one copy, they won't develop cystic fibrosis. However, they will be carriers and could pass the gene to their own children.


# Causes

- ▶ In cystic fibrosis, a defect (mutation) in a gene — the cystic fibrosis transmembrane conductance regulator (CFTR) gene — changes a protein that regulates the movement of salt in and out of cells. The result is thick, sticky mucus in the respiratory, digestive and reproductive systems, as well as increased salt in sweat.
- ▶ The CFTR gene provides instructions for making a protein called the CF transmembrane conductance regulator (CFTR). This protein functions as a channel across the membrane of cells that produce mucus, sweat, saliva, tears, and digestive enzymes. The channel transports negatively charged particles called chloride ions into and out of cells. The transport of chloride ions helps control the movement of water in tissues, which is necessary for the production of thin, freely flowing mucus. Mucus is a slippery substance that lubricates and protects the lining of the airways, digestive system, reproductive system, and other organs and tissues.
- ▶ Many different defects can occur in the gene. The type of gene mutation is associated with the severity of the condition.

# Types of CF mutations

- There are five classes of CFTR mutations: protein production, protein processing, gating, conduction, and insufficient protein.
- More than 2,000 different mutations of the CF gene have been identified. The most common CF mutation, F508del, is primarily considered to be a protein processing mutation.





# Early childhood Symptoms and diagnosis

- ▶ Sweat glands offer an interesting contrast to all other tissues containing CFTR channels in that the flow of chloride is reversed. Normally, sweat glands move chloride from the extracellular space into the intracellular space. Thus, sodium and water are reabsorbed from the sweat gland tissues into the body. However, failure of the chloride channel to reabsorb chloride leads to a loss of sodium onto the skin surface and a subsequent fluid loss. This causes the pathognomonic salty skin seen with cystic fibrosis.



# Respiratory signs and symptoms

- ▶ The thick and sticky mucus associated with cystic fibrosis clogs the tubes that carry air in and out of the lungs. This can cause signs and symptoms such as:
  - ▶ A persistent cough that produces thick mucus (sputum)
  - ▶ Wheezing
  - ▶ Exercise intolerance
  - ▶ Repeated lung infections
  - ▶ Inflamed nasal passages or a stuffy nose
  - ▶ Recurrent sinusitis





# Digestive signs and symptoms

- ▶ The thick mucus can also block tubes that carry digestive enzymes from the pancreas to the small intestine. Without these digestive enzymes, the intestines aren't able to completely absorb the nutrients in the food. The result is often:
- ▶ Foul-smelling, greasy stools
- ▶ Poor weight gain and growth
- ▶ Intestinal blockage, particularly in newborns (meconium ileus)
- ▶ Chronic or severe constipation, which may include frequent straining while trying to pass stool, eventually causing part of the rectum to protrude outside the anus (rectal prolapse)



# Reproductive system

- ▶ In the reproductive system, the thickened secretions can cause obstructions and affect the development and function of the sexual organs. Most males with CF have obstruction of the sperm canal known as congenital bilateral absence of the vas deferens (CBAVD).



Thank you for listening