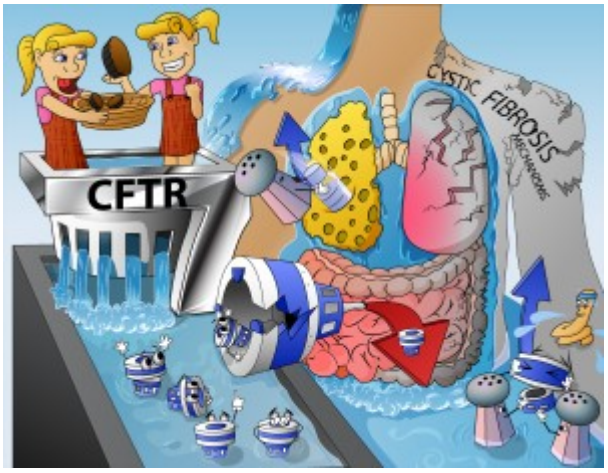
Cystic Fibrosis Mechanisms

Cystic fibrosis is a hereditary disease leading to problems with Cl- channels in the body. It is the most common lethal genetic disease in the Caucasion population. Patients develop recurrent pulmonary infections, bronchitis, infertility, pancreatic insufficiency, steatorrhea and malabsorption.



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Autosomal Recessive
Recessive-chocolate
Cystic fibrosis is inherited in an autosomal recessive manner.

CFTR Chromosome 7
CFTR-sifter Chrome 7
This disease occurs due to a defect in the CFTR gene on chromosome 7. Though there are many ways to effect the CFTR gene, commonly, a deletion of Phe508 occurs.

Cl- channel Defect
Chlorine-dispenser Channel Broken
CFTR encodes an ATP-gated Cl- channel. In the lungs and gut, this channel secretes Cl-, which leads to an H2O gradient. In sweat glands, this channel reabsorbs Cl-. A defect in CFTR leads to defects in Cl- secretion through these channels.

Decreased Chloride Secretion
Down-arrow Chlorine-dispenser Secreting into GI and Lungs
Defective Cl- channels lead to increased chloride on the skin (not reabsorbed in sweat glands), as well as decreased chloride secretion (and subsequently water) in the gut and lungs.

Increased Na and Water Reabsorption
Up-arrow Salt-shaker and Water-bottle pulled out of Absorbing-sponge
In this disorder, Cl- is not secreted into the lungs and GI tract. Thus, there is increased intracellular Cl-, which then causes a compensatory increase in Na+ reabsorption. Due to the high concentration of accumulated salt (NaCl) intracellularly, water is then reabsorbed.

Increased Na and Cl in Sweat
Up-arrow Salt-shaker and Chorine-dispenser at Sweaty-sweatgland
In this disorder, Cl- is not reabsorbed through sweat glands. Increased epithelial Cl- causes a compensatory increase in Na+ excretion via epithelial channels.

Dehydration of Mucous Layers
Dried-up Mucous Layers of body
As Cl- is not secreted into the lungs and GI tract and is "trapped" intracellularly, Na+ follows and H2O is reabsorbed. This leads to abnormally thick mucus secreted into the lungs and GI tract.