

## Cystic Fibrosis Mechanisms

Cystic fibrosis is a hereditary disease leading to problems with Cl<sup>-</sup> channels in the body. It is the most common lethal genetic disease in the Caucasian 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<sup>-</sup> channel Defect

#### Chlorine-dispenser Channel Broken

CFTR encodes an ATP-gated Cl<sup>-</sup> channel. In the lungs and gut, this channel secretes Cl<sup>-</sup>, which leads to an H<sub>2</sub>O gradient. In sweat glands, this channel reabsorbs Cl<sup>-</sup>. A defect in CFTR leads to defects in Cl<sup>-</sup> secretion through these channels.

### Decreased Chloride Secretion

#### Down-arrow Chlorine-dispenser Secreting into GI and Lungs

Defective Cl<sup>-</sup> 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<sup>-</sup> is not secreted into the lungs and GI tract. Thus, there is increased intracellular Cl<sup>-</sup>, which then causes a compensatory increase in Na<sup>+</sup> 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 Chlorine-dispenser at Sweaty-sweatgland

In this disorder, Cl<sup>-</sup> is not reabsorbed through sweat glands. Increased epithelial Cl<sup>-</sup> causes a compensatory increase in Na<sup>+</sup> excretion via epithelial channels.

**Dehydration of Mucous Layers**

**Dried-up Mucous Layers of body**

As Cl<sup>-</sup> is not secreted into the lungs and GI tract and is "trapped" intracellularly, Na<sup>+</sup> follows and H<sub>2</sub>O is reabsorbed. This leads to abnormally thick mucus secreted into the lungs and GI tract.