

## Cystic Fibrosis Mechanisms

Cystic fibrosis is a hereditary disease leading to problems with  $\text{Cl}^-$  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.

### $\text{Cl}^-$ channel Defect

#### Chlorine-dispenser Channel Broken

CFTR encodes an ATP-gated  $\text{Cl}^-$  channel. In the lungs and gut, this channel secretes  $\text{Cl}^-$ , which leads to an  $\text{H}_2\text{O}$  gradient. In sweat glands, this channel reabsorbs  $\text{Cl}^-$ . A defect in CFTR leads to defects in  $\text{Cl}^-$  secretion through these channels.

### Decreased Chloride Secretion

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

Defective  $\text{Cl}^-$  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,  $\text{Cl}^-$  is not secreted into the lungs and GI tract. Thus, there is increased intracellular  $\text{Cl}^-$ , which then causes a compensatory increase in  $\text{Na}^+$  reabsorption. Due to the high concentration of accumulated salt ( $\text{NaCl}$ ) intracellularly, water is then reabsorbed.

### Increased Na and $\text{Cl}^-$ in Sweat

#### Up-arrow Salt-shaker and Chlorine-dispenser at Sweaty-sweatgland

In this disorder,  $\text{Cl}^-$  is not reabsorbed through sweat glands. Increased epithelial  $\text{Cl}^-$  causes a compensatory increase in  $\text{Na}^+$  excretion via epithelial channels.

## Dehydration of Mucous Layers

### Dried-up Mucous Layers of body

As  $\text{Cl}^-$  is not secreted into the lungs and GI tract and is "trapped" intracellularly,  $\text{Na}^+$  follows and  $\text{H}_2\text{O}$  is reabsorbed. This leads to abnormally thick mucus secreted into the lungs and GI tract.