The Biology of CF

CF is caused by a defective or missing cystic fibrosis transmembrane conductance regulator (CFTR) protein resulting from mutations in the CFTR gene.

NORMAL CFTR PROTEINS
lie on the surface of cells that line the lungs, digestive system and sweat glands, and are responsible for regulating the passage of salt and water into and out of cells.

DEFECTIVE CFTR PROTEINS
result in poor flow of salt and water into and out of cells.

THE RESULT
Abnormally sticky mucus forms and obstructs the lungs and airways, intestine, and the ducts of the pancreas.

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How CF affects the body

LUNGS
Mucus buildup in the lungs causes chronic lung infections and progressive lung damage. Half of all lung function decline in people with CF is associated with pulmonary exacerbations, or episodes of worsening of respiratory symptoms due to lung infections. Pulmonary exacerbations can lead to permanent reduction in lung function, and lung disease is the leading cause of death of people with CF.

LIVER
Thicker bile and blocked bile ducts can lead to cirrhosis, making it hard for the liver to work properly.

GASTROINTESTINAL TRACT
Reduced ability to absorb nutrients can make it difficult to maintain a healthy weight.

SINUSES
Chronic congestions, sinus infections, and nasal polyps may require treatment with medication and/or surgery.

SWEAT
Excessive salt can cause dehydration and overheating.

PANCREAS
Decrease in pancreatic enzymes leads to malnutrition. Meanwhile, because the pancreas makes too little insulin, and does not properly absorb insulin, some people with CF may develop cystic fibrosis-related diabetes.

REPRODUCTIVE SYSTEM
Most males with CF are infertile; and many females experience reduced fertility.