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Cystic fibrosis

Updated: January 31, 2011.

Cystic fibrosis (CF) is the most common fatal genetic disease in the United States today. It causes the body to produce a thick, sticky mucus that clogs the lungs, leading to infection, and blocks the pancreas, stopping digestive enzymes from reaching the intestines where they are required to digest food.

CF is caused by a defective gene, which codes for a chloride transporter found on the surface of the epithelial cells that line the lungs and other organs. Several hundred mutations have been found in this gene, all of which result in defective transport of chloride, and secondarily sodium, by epithelial cells. As a result, the amount of sodium chloride (salt) is increased in bodily secretions. The severity of the disease symptoms of CF is directly related to the characteristic effects of the particular mutation(s) that have been inherited by the sufferer.

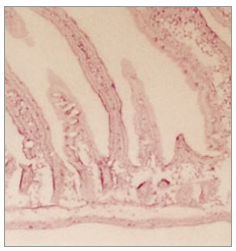
CF research has accelerated sharply since the discovery of CFTR in 1989. In 1990, scientists successfully cloned the normal gene and added it to CF cells in the laboratory, which corrected the defective chloride transport mechanism. This technique—gene therapy—was then tried on a limited number of CF patients. However, this treatment may not be as successful as originally hoped. Further research will be required before gene therapy, and other experimental treatments, prove useful in combating CF.

Related diseases

See other Diseases of the Digestive System

See other Neonatal Diseases

See other Respiratory Diseases



Building mouse models of human disease. Expression of a human cystic fibrosis (CFTR) gene in the gut of a mouse. A human antisense probe was used to show human CFTR expressed in the mouse duodenum.

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