Cystic Fibrosis (CFTR)

Cystic Fibrosis is caused by defects in the CFTR gene. The gene encodes for the protein cystic fibrosis transmembrane conductance regulator, which is involved in chloride ion transport and hence mucus production. Sufferers have breathing difficulties, due to sticky mucus buildup in the lungs. They also have severe digestive problems, since pancreatic enzymes are blocked from entering the small intestine, the main locus of nutrient absorption. Male sufferers are normally infertile, due to the lack of functioning vas deferens tubes which lead to the urethra. Diabetes and liver disease are common complications that often develop over time. The mean projected survival time for children born with the condition in the USA in 2010 was estimated at 37 years for women and 40 years for men.

The inheritance of the defective gene is autosomal recessive, typically requiring both parents to be asymptomatic carriers of the mutated gene. The condition is relatively common among Caucasian Americans, with an incidence of about 1 in 2,500 to 1 in 3,500. The figures are about 1 in 4,000 to 10,000 for Hispanic Americans, 1 in 15,000 to 20,000 for African Americans, and 1 in 100,000 for Asian Americans. It is estimated that 1 in 29 Caucasian Americans carry the defective gene asymptomatically. The figures are 1 in 46 for Hispanic Americans, 1 in 65 for African Americans, and 1 in 90 for Asian Americans.

Sources

Cystic Fibrosis Foundation, Carrier Testing for CF.
See http://www.cff.org/AboutCF/Testing/Genetics/GeneticCarrierTest/

MacKenzie, T. *et al.* (2014), “Longevity of Patients with Cystic Fibrosis in 2000 to 2010 and Beyond: Survival Analysis of the Cystic Fibrosis Foundation Patient Registry,” *Annals of Internal Medicine,* 161, 233-241.

NIH, Genetics Home Reference: Cystic Fibrosis.
See http://ghr.nlm.nih.gov/condition/cystic-fibrosis