

**Disease Name:**

# Cystic Fibrosis

**Classification:** Channelopathy

**Genetic Information:**

**Inheritance:** Autosomal recessive

**Population Incidence:** 1:3,000

**Ethnic Incidence:** Caucasian (non-Hispanic) and Askenazi Jews more frequent, but CF occurs in all races.

**Gene & Location:** Cystic fibrosis conductance regulator gene; 7q31.2

**Common Mutation:** Delta-F508 in 70% of Caucasians; (over 1000 mutations known)

**OMIM #** #219700

**Disease Information:**

**Symptom Onset:** 20% as neonates with meconium ileus; the rest in childhood or adulthood.

**Symptoms:** CF affects salt transport across cells that line the sweat glands, pancreas, liver, lungs and reproductive tract. 95% of CF individuals have some degree of pancreatic insufficiency. This leads to digestion problems, failure to thrive and frequent foul smelling stools. Thick sticky mucous leads to chronic cough, recurrent lung infections, wheezing or other lung symptoms.

**Physical Findings:** There may be no symptoms in the newborn. Pancreatic insufficiency leads to failure to thrive, malnutrition and vitamin deficiencies. Lung involvement includes chronic cough, recurrent infections and diminishing lung capacity. Infertility occurs in 95% of males. Women may have reduced fertility, but successful pregnancy and normal offspring have been reported.

**Treatment:** Enzyme replacement if pancreas is insufficient; high calorie, high protein diet, lung physiotherapy and organ transplant.

**Natural History without Treatment** Progressive failure to thrive and lung disease results in death in early to mid-childhood.

**Natural History with Treatment** Comprehensive care can result in survival into the third decade.

**Metabolic  
Information:**

**IRT Results:**

>100 ng/mL First Newborn Screen, >80 ng/mL Second Newborn Screen





**Prenatal testing:**

Amniotic fluid, chorionic villus, DNA testing available

**Miscellaneous  
Information:**

Prepared for the NW Regional Newborn Screening Program by Michael Wall at Oregon Health & Science University.

**References:**

1. Cystic Fibrosis Foundation web site: [www.cff.org](http://www.cff.org)
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3. [Davies JC. Related Articles, Links](#)  Gene and cell therapy for cystic fibrosis. Paediatr Respir Rev. 2006;7 Suppl 1:S163-5. Epub 2006 Jun 6.
4. [Li L, Zhou Y, Bell CJ, Earley MC, Hannon WH, Mei JV. Related Articles, Links](#)  Development and characterization of dried blood spot materials for the measurement of immunoreactive trypsinogen. J Med Screen. 2006;13(2):79-84.
5. [Dodge JA, Turck D. Related Articles, Links](#)  Cystic fibrosis: Nutritional consequences and management. Best Pract Res Clin Gastroenterol. 2006 Jun;20(3):531-46.