
Genetic Services Policy Project

Cystic Fibrosis

What are the symptoms of Cystic Fibrosis (CF)?

- Patients with CF produce mucus that clogs the lungs and results in lung infections.
- Patients with CF have difficulty in absorbing nutrients from food because the mucus keeps digestive enzymes from reaching the intestines.
- Liver damage and hepatic failure may result from blocked bile ducts.
- Median life expectancy for individuals with CF has increased from under 10 years old in the 1960s to 33.4 years.

Who is affected by CF?

- CF affects approximately 30,000 children and adults in the U.S. and occurs in approximately 1 in 3,500 live births.
- CF most often affects people of European descent.
- 80% of patients with CF are diagnosed by age three; only 10% of patients are diagnosed after age 18.

What causes CF?

- CF is a genetic condition caused by an autosomal recessive mutation.
- The estimated carrier frequency for a mutation is between 1 in 25 and 1 in 29 among European-Americans.
- The $\Delta F508$ mutation, the most common gene variant, accounts for about 70% of CF carriers.
- Researchers have identified over 1,000 mutations.
- Non-genetic modifiers, such as environmental tobacco smoke, respiratory pathogens, and socioeconomic status (SES), may impact health outcomes.

How is CF detected?

- Carrier and prenatal screening: Expert opinion has recommended genetic testing for CF for adults with a positive family history, partners of people with CF, couples planning a pregnancy, and couples seeking prenatal care.
- Newborn screening: Inclusion of CF on newborn screening panels has been suggested and endorsed by the Centers for Disease Control and Prevention because of potential benefits from early nutritional treatment, such as improved growth.
- Diagnosis in childhood and adulthood.

What are standard treatments and therapies for CF?

- Treatment depends on the stage of disease and the organs involved.

- Chest physical therapy involves clapping on the back and chest to dislodge mucus.
- Antibiotic treatments like TOBI® (tobramycin) aerosolized antibiotic help to address lung infections. Azithromycin is another antibiotic used for patients who are chronically infected with *Pseudomonas aeruginosa* bacteria.
- Pulmozyme® is a mucus-thinning drug for improving lung function.
- Enzyme replacement is a therapy for pancreatic insufficiency.
- To combat nutritional deficiencies, patients may also take nutritional supplements.
- New therapies for CF include recombinant human DNase (rhDNase; dornase alfa) treatment.
- Lung transplantation and lifetime use of antirejection medication and immunosuppressant therapies treat patients with end-stage CF.

What are costs associated with CF?

- Lifetime direct costs of CF are estimated at \$200,000 to \$300,000 (1996 values, 5% discount rate).
- Annual cost of medical care in 1996 averaged \$13,300 per patient, ranging from \$6,200 among patients with mild disease to \$43,300 among patients with severe disease
- Of total costs, 47% were from hospitalization, 18% were from DNase (Pulmozyme), 12% were from clinic visits, and 10% were from outpatient antibiotics.

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