FACT SHEET

# Cystic Fibrosis

NATIONAL INSTITUTES OF HEALTH

## Yesterday

- Cystic fibrosis (CF), an inherited disease of the mucus and sweat glands, affects the lungs, pancreas, liver, intestines, sinuses, and sex organs.
- CF was easily diagnosed with a "sweat" test but physicians had few therapies to thin the mucus build-up that occurs in the lungs and digestive tracts of people with the disease.
- Doctors had no effective antibiotics for treating the infections, especially those caused by the bacterium Pseudomonas aeruginosa, which thrive in the mucus in CF patients' lungs and gradually damage lung tissue.
- Digestive and pancreatic manifestations of the disease frequently resulted in delayed growth and malnutrition.
- In 1962, the predicted median survival for CF patients was about 10 years, with few surviving into their teenage years.
- NIH researchers discovered that the underlying problem in patients with CF was a defect in salt transport through their cell membranes.
- The CF transmembrane conductance regulator (CFTR) gene, which is defective in people with CF, was discovered in 1989.
- Carrier detection and prenatal diagnosis of CF were developed for families with a history of the disease.
- Mouse models of the disease were developed in the early 1990s, but unlike humans, mice without CFTR did not exhibit typical symptoms of CF, including the lung and liver disease that cause the premature mortality seen in humans.

## Today

- Wide-ranging research on CF resulted in dramatic improvements in the treatment of the disease, almost quadrupling the median life expectancy to 37 years of age in the U.S. In parts of the world where state-of-the-art medical care is not available, life expectancy remains much lower.
- Most states conduct newborn screening for CF so that, even before symptoms appear, treatment can be initiated to prevent malnutrition and growth delay.
- Powerful new antibiotics, such as inhaled tobramycin, are much more effective than older drugs for treating Pseudomonas aeruginosa and other devastating bacterial lung infections that frequently develop.
- Other drugs, such as Pulmozyme® and azithromycin, slow the progression of the lung disease associated with CF.
- New mechanical chest physical therapy devices help loosen the mucus and make it easier for patients to clear their lungs.
- Lung transplantation has become an option for some people with CF who have severe lung damage.
- The digestive problems in people with CF can be managed with nutritional therapy, oral pancreatic enzymes, and medications to reduce stomach acid.
- Diabetes in people with CF is now recognized, and effective diabetes therapy is available to improve their health.
- A pig model for CF was developed that may provide a powerful tool for testing new therapeutic strategies. Newborn piglets that lack CFTR have signs similar to those seen in human infants with CF, including abnormalities in the intestines, pancreas, and liver.
- Treatment for CF has greatly extended life expectancy and improved quality of life, but there is no cure. Effective treatment still requires hours of demanding daily therapy.

## Tomorrow

- New therapeutics are currently in development, some of which may provide a functional CFTR protein in patients with some versions of the gene, potentially

eliminating many disease complications and allowing people with the disease to live essentially normal lives.

- Other new therapies may dramatically improve the salt–water balance in patients with CF, allowing them to clear mucus from their lungs and experience fewer infections.
- Gene therapy—insertion of a healthy CFTR gene to act in place of the defective version of the gene—could effectively cure the disease.
- Identification of modifier genes may be used to predict patients who are likely to have more severe lung disease. For example, a gene with variants producing high levels of mannose binding lectin 1 helps protect against CF-related lung infections, while a genetic variation leading to high levels of TGF-beta protein has the opposite effect.

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