



M3 GLOBAL RESEARCH REVIEW

CYSTIC FIBROSIS

WHAT IS CYSTIC FIBROSIS

Cystic fibrosis (CF) is an inherited disease that is usually diagnosed at an early age. As recently as the 1980's, most patients could not expect to live into adulthood. The disease arises when a patient has two defective copies of the cystic fibrosis transmembrane conduct regulator (CFTR) gene, first isolated in 1989. The mutated gene underperforms, alters membrane fluid transport, and causes mucus to accumulate in the lungs and other organs. Clearing the mucus from the lungs via both physical and medicinal means was the mainstay of CF treatment strategies for many years. Many patients also suffer severe digestive issues from compromised pancreatic function, and death from pulmonary infections was a common outcome.

CYSTIC FIBROSIS FOUNDATION

According to WHO, this inheritable disease affects less than 80,000 patients worldwide, but is likely under-diagnosed in Latin America, Africa, and India. Caucasian populations have the highest incidence and prevalence. The Cystic Fibrosis Foundation Patient Registry estimates that there are roughly 1,000 newly diagnosed cases per year and approximately 30,000 patients in the US have CF.

The Cystic Fibrosis Foundation also estimates that as many as 10 million people may carry a single copy of the defective gene. The American Lung Association says CF is the 2nd most common inherited disorder in children in the US. More than half of the CF population is 18 years or older, with an median survival age of almost 40 years. This is a dramatic improvement from the 1950's when a child with CF was likely to die before age 5.

The Cystic Fibrosis Foundation (CFF) not only serves to disseminate CF information, but is extremely active in focusing clinical research on new therapies for patients with CF. They are an important supporter of CF clinical trials with 10 CFF Research Centers and more than 120 CF care centers in the US. Most US trials in patients with CF are conducted with involvement of the Foundation. US guidelines for the diagnosis of CF were recently revised to include CFTR2 gene testing, standardized diagnostic criteria for diagnostic tests conducted outside of newborn screening, and lower the threshold of sweat chloride concentrations for a "possible" CF or related disease diagnosis.

AWARENESS

May is Cystic Fibrosis Awareness Month. Over 500 events are held each year in the US, with more than 125,000 participants last year.

The CF disease area has been a primary beneficiary of the advances in human genome testing. The CFTR gene mutations as the genetic base cause was discovered in 1989. There are roughly 2,000 known mutations in CFTR, with 300 known to cause the disease. Two of those mutations have responded to new therapies that improve chloride ion transport and decrease lung mucus accumulation. Pulmonologists and pediatricians routinely perform CFTR screening for new CF patients to identify those who can benefit from these new therapies. Work continues to discover therapies for other known mutations, and improve efficacy for patients with these two mutations.

THE COMPLETE PULMONOLOGIST

M3 is active in recruiting for our client's studies in the pulmonary and pediatric disease area. In 2016, we recruited over 14,000 respondents for >220 studies with >150 clients across 67 countries. Most (143) were online quantitative surveys, 63 were qualitative only projects, and 16 included both study type activities. Cystic Fibrosis was the primary topic area in 9 studies among >725 respondents in 12 countries.

M3 also provides several syndicated services in this sector. *The Complete Pulmonologist, Complete Pediatrician, and Complete Gastroenterologist* are syndicated studies examining the challenges faced by specialists in their daily practice. *Physician Map* is a KOL mapping service offered in specific disease areas. *Patient Map* is a 5,000 respondent syndicated service that examines patient loads across 400 diseases.

Contact us on how to access our robust panel of Pulmonologist or for additional information on our syndicated offerings.

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