

Cystic Fibrosis

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Definition: Cystic Fibrosis (CF) is the second most common life shortening, early onset inherited disorder in the US, after Sickle Cell Anemia. About 30,000 individuals in the US have CF (2). CF causes thick, sticky mucus to build up in the lungs and digestive system and other organs of the body. This mucus frequently obstructs the lungs, leading to progressive chronic and life-threatening lung infections, and the pancreatic ducts, preventing normal digestion and leading to malabsorption and malnutrition.

Outcomes without screening: The most common symptoms of CF are salty tasting skin, persistent coughing, wheezing, shortness of breath, recurring pneumonia, an excessive appetite but poor weight gain and greasy, bulky stools. Most individuals with CF are not diagnosed until they show symptoms of the disease over several years.

Incidence: CF occurs most commonly among whites. It is estimated that 1/2,500 to 3,200 non Hispanic white births are affected in comparison to 1/10,500 Native Americans, 1/11,500 Hispanics, 1/14,500 African Americans, and 1/25,500 Asians. More than 10 million Americans are asymptomatic carriers of a defective CF gene.

Outcomes with screening: The first step of newborn screening is to identify all infants with elevated initial and repeat Trypsinogen levels. The next step is to confirm CF by sweat test and begin treatment. Early diagnosis allows for immediate intervention with specialized therapies, including pancreatic enzymes to aid digestion and a high calorie, high fat diet. These interventions improve height, weight and cognition; they also may help maintain respiratory function, increase life expectancy and reduce hospitalizations. Newborn screening for CF enables treatment before symptoms occur, which can improve nutrition and decrease complications, giving newborns the best chance for a healthier future.

Newborn screening for CF is still important even if parents' carrier screening tests were negative. CF carrier screening only identifies the most common of the more than 1,400 CF gene mutations. Therefore, it is still important that all newborns be given the newborn screening test for CF.

Cause of CF: To have CF an individual must inherit two defective CF genes - one from each parent. Each time two CF carriers conceive there is a 25 percent chance that their child will have CF; a 50 percent chance that the child will be a CF carrier; and a 25 percent chance that the child will not be a CF carrier.

Screening test and confirmation: Recently, the Centers for Disease Control and Prevention (CDC) issued a recommendation that all states consider routine newborn screening for CF. In 2000, about 400,000 children born in the US were screened for CF and this was expected to increase to 800,000 by the end of 2004.

The CF newborn screening test is not a diagnostic test. Babies are screened for CF before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of Trypsinogen. Since only a fraction of babies with an initial positive Trypsinogen screening test will actually have CF, the screening test is

repeated. If both Trypsinogen screening tests are elevated a confirmatory sweat test is done to rule out or confirm a CF diagnosis.

Treatment: The treatment of CF depends upon the severity of symptoms and the organs involved. Most people with CF must take pancreatic enzyme supplements with every meal to absorb enough calories and nutrients to grow and stay healthy. They also must eat a high-calorie, high fat diet. People with CF also perform daily airway clearance therapy to help clear mucus from the lungs. Other types of treatments include antibiotics to fight lung infections and drugs to thin the mucus and improve lung function.

Advances in CF care since the early 1980s have contributed to substantial improvements in measures of malnutrition, lung function, and mortality among children and adolescents with CF. During 1985-2002, predicted survival of persons with CF in the United States increased from age 25 years to age 33 years.

Special concerns and issues: For a disease to be included in newborn screening test panels, it should meet certain guidelines. These include benefit of early diagnosis; availability of a confirmatory test; and improved health because of early detection and timely treatments. The benefits of newborn screening for CF have been documented and studied extensively. New treatments and specialized care for CF improve and extend the lives of people with CF. In addition, early diagnosis and proper care of babies with CF can have significant impact on their nutritional status and allows them to grow and develop to their genetic potential.

References:

1. <http://www.cdc.gov/mmwr/preview/mmwrhtml/rr5313a1.htm>
2. http://www.cff.org/UploadedFiles/living_with_cf/Files/NBSFinalQALH.pdf
3. <http://www.lungusa.org/site/pp.asp?c=dvLUK9O0E&b=35042>