Treatment of Cystic Fibrosis

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Key Points
➢ CF – an inherited, autosomal recessive disorder ➢ CF patients face a variety of medical problems ➢ Caused by a mutation in the CFTR gene ➢ Diagnosis is by sweat and genetic tests

Cystic fibrosis is an inherited autosomal recessive disorder, its main cause is mutations in CFTR gene, CFTR is a transmembrane conductance regulator gene. People having Cystic fibrosis face a wide variety of medical conditions which affect the endocrine, pulmonary, gastrointestinal, biliary, pancreatic, and reproductive systems. In past carrier of CF having one defective copy of CFTR, were not thought to be at risk for CF-associated diseases. But now a days recent studies suggests that heterozygotes are at increased risk for many of the same conditions as homozygotes. Like heterozygotes appear to be at increased risk for atypical mycobacterial infections, chronic pancreatitis, and bronchiectasis. There are almost 10 million CF carriers in the United States. The new techniques like Universal newborn screening and prenatal genetic screening will identify more carriers of CF. Thus, it is essential to develop more precise estimates of health risks attributable to the Cystic fibrosis carrier state across the lifespan.[1]

Symptoms:

In the United States due to newborn screening, cystic fibrosis can be diagnosed before symptoms develop, within the first month of life. But people born before newborn screening, Cystic fibrosis in them may not be diagnosed until the signs and symptoms of CF appear. The signs and symptoms of Cystic fibrosis vary, rely on the severity of the disease. Even in the same person, as the time passes symptoms may worsen or improve. Some people may not experience any symptoms of the disease until their teenage years or adulthood. People who are not diagnosed until adulthood ,usually these people have milder disease and are more likely to have atypical symptoms, such as infertility and recurring pneumonia, recurring bouts of an inflamed pancreas (pancreatitis).The patients of cystic fibrosis have a higher than normal level of salt in their sweat. When the parents kiss their children, they can often taste salt. The other signs and symptoms of CF affect the digestive system and respiratory system.

Respiratory signs and symptoms

In Cystic fibrosis the thick and sticky mucus clogs the tubes that carry air in and out of lungs. It can cause signs and symptoms like: A persistent cough which produces thick mucus (sputum), repeated lung-infections exercise intolerance, wheezing, inflamed nasal passages, recurrent sinusitis, or a stuffy nose.

Digestive signs and symptoms

The thick mucus also blocks the tubes which carry digestive enzymes from pancreas to small intestine. Intestine cannot absorb the nutrients from the eaten food without the help of these digestive enzymes. It may lead to: foul-smelling, greasy stools, poor weight gain and poor growth. Intestinal blockage due to secretion of mucous, particularly in newborns (meconium ileus) .Severe or chronic constipation, which may include repeated straining while trying to pass stool, by and by causing part of the rectum to protrude outside the anus.[2]

Causes:

Cystic fibrosis (CF) is a monogenic disease which is caused by mutations in the CFTR gene on the chromosome 7, It is a complex and greatly variable in clinical expression. In this disease pancreas, airways, male genital system, intestine, bone, liver, and kidney are involved. The lack of CFTR or its malfunctioning causes fat mal-absorption and chronic pulmonary infections leading to progressive lung bronchiectasis and progressive lung damage.[2] Beyond lower airway disease, cystic fibrosis has obvious impact on other organ systems.[3]

Diagnostic Tests of Cystic Fibrosis:

Sweat Test which measures the amount of chloride in the sweat. Genetic Test which detects chromosomal mutations associated with disease.

Treatment:

Gene therapy is the advanced treatment for cystic fibrosis (CF). It involves the transfer of correct copies of cystic fibrosis transmembrane conductance regulator (*CFTR*) DNA in the epithelial cells of airways.[4] Trikafta pills are also used in the treatment of cystic fibrosis. Bronchodilators, hypertonic saline, Dornase alfam, aerosolized antibiotic are the medications used to treat cystic fibrosis.[4]

References:

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