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Opinion Article

Diagnosis and Treatment Methods for Cystic Fibrosis

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DESCRIPTION

Cystic fibrosis (CF) is a rare genetic disorder that mostly affects the lungs but also affects the pancreas, liver, kidneys, and gut. Long-term consequences of repeated lung infections include difficulty breathing and coughing up mucus. Sinus infections, poor growth, fatty stool, clubbing of the fingers and toes, and infertility in most males are further signs and symptoms. Different persons may experience varying degrees of symptoms.

Cystic fibrosis is an autosomal recessive disorder. It is caused by mutations of the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) protein gene. Those who have a single functional copy are carriers and generally healthy. Sweat, digestive fluids, and mucus are all produced by CFTR. When the CFTR is not working properly, normally thin secretions thicken.

DIAGNOSIS

Newborn screening

Newborn screening has grown standard over the previous decade and is currently accessible in all US states. The newborn screening detects infants with high levels of an enzyme called immunoreactive trypsin in their blood. This happens when the pancreas is injured. If the results are abnormal, the test is repeated. Some states also combine this with testing for deltaF508, the most prevalent gene mutation. Because there are numerous false-positive tests, the infant will be referred for more testing. This comprises collecting a blood sample and/or performing a sweat test to determine whether the newborn has two genes that cause CF.

Genetic testing

The CF gene has been found to contain over 2,000 distinct mutations. The majority of them are extremely rare, but a few are quite prevalent, such as the deltaF508 mutation, which is seen in at least 70% of people with CF. In most situations, genetic testing can pinpoint the particular mutation. Genetic testing is especially crucial for couples who wish to have children because more than 10 million Americans are carriers of a CF gene. When both parents are carriers, there is a one-in-four risk that the child will have CF.

Sweat test

Sweat is collected from a tiny region on the child's forearm and chloride levels are determined. Children with cystic fibrosis have excessive chloride levels in their sweat because a deficiency of CFTR prevents salt on the skin from being reabsorbed into the sweat glands.

Measuring nasal lining

A tiny electrical current across the nasal lining might also be used to confirm the diagnosis (epithelium). The nasal lining is treated with various solutions, and the electrical current is monitored. This test produces considerably different results in people with CF than in people without CF, and it may help confirm a diagnosis.

TREATMENT METHODS

Airway clearance therapy

Airway clearance therapy is required for people with cystic fibrosis. This can be accomplished through manual chest physical therapy or by wearing a device that helps clear airway secretions by shaking the mucus in the airways, allowing you to cough it up. Breathing through a mask or mouthpiece is another portable mucus clearing technique. This device forces you to breathe out more forcefully, dislodging mucus that has been lodged in your airways.

Enzymes and nutrients

Pancreatic enzyme replacement therapies aid the body's absorption of meals and nutrients. Enzymes must be taken

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before each meal or snack. People with CF must additionally take vitamins that are absorbed with fat. Additional salt should be included in the formula or meal.

Lung transplants

A lung transplant may be advised in severe cases of cystic fibrosis when the lungs stop performing correctly and all medicinal treatments have failed. A lung transplant is a risky procedure, but it can significantly enhance the length and quality of life for those with severe cystic fibrosis.