Commentary

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Diagnosis and Treatment of Cystic Fibrosis

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Introduction

Cystic fibrosis (CF) is a genetic disease that causes sticky and thick mucus build-up in organs like the lungs, digestive system, and other body organs severely. This thick mucus clogs the airways and making it difficult to breathe.

Mucus is a slick substance that coats the linings of the lungs, digestive system, reproductive system, and other organs and tissues, lubricating and protecting them. The body creates extremely thick and sticky mucus in patients with cystic fibrosis. This abnormal mucus can clog the airways, causing serious breathing difficulties as well as bacterial infections in the lungs. Coughing, wheezing, and inflammation are all symptoms of these infections. Over time, mucus build-up and infections cause long-term lung damage, including scar tissue (fibrosis) and cysts in the lungs.

Causes

Cystic fibrosis is a hereditary disease. CF patients inherit two defective genes, one from each parent. CF is recessive because it requires two gene variants to be diagnosed with the disease. (Gene mutation is an earlier term for gene variation).

You don't have to have cystic fibrosis if your parents do. In truth, many families do not have a history of cystic fibrosis. If no one in your family has ever had cystic fibrosis, the person who has the gene mutation is known as a carrier. In the United States, about 1 in 31 people is a carrier who does not have CF symptoms.

Symptoms

1. Respiratory signs and symptoms

Mucus from cystic fibrosis is thick and sticky, and it clogs the airways in and out of the lungs. This can result in the following signs and symptoms:

- A cough that lasts for a long time and generates a lot of mucous (sputum)
- Wheezing
- Intolerance to exercise
- Lung infections that recur
- A stuffy nose or inflamed nasal passages
- Sinusitis that recurs

2. Digestive signs and symptoms

The thick mucus can also clog digestive enzyme tubes that run from your pancreas to your small intestine. Your intestines won't be able to absorb all of the nutrients in the food you eat if you don't have these digestive enzymes. Frequently, the end outcome is:

- Stools with a foul odour and a greasy consistency
- Weight gain and growth are both poor.
- Blockage of the intestines, especially in neonates (meconium ileus)
- Constipation that is chronic or severe, with continuous straining while attempting to pass stool, eventually causes a portion of the rectum to protrude outside the anus (rectal prolapse).

Diagnosis

- Blood test: this test is done to determine the immunoreactive trypsinogen (It). People with CF have higher levels of IT in the blood.
- DNA test: This check for any mutations to the CFTR gene.
- New born screening: Usually a healthcare worker collects a few drops of blood from a heel prick and inserts them on a special card called a Guthrie card, which is normally done while you're new-born, is in the hospital. The exam screens for a variety of illnesses, including cystic fibrosis. Every state in the United States requires neonates to be tested at birth and again a few weeks later.
- Sweat test: A perspiration test determines how much chloride is present in the body's sweat, which is higher in persons with CF.
- Lung function tests: A spirometer is used to perform the most common lung function test. You take a deep breath in and then push the inhaled air into the spirometer's mouthpiece.

Treatments

Although cystic fibrosis has no cure, medication can help to alleviate symptoms, prevent complications, and enhance quality of life. Close supervision and early management are indicated to slow the progression of CF, which can lead to longevity. Because cystic fibrosis treatment is difficult, you should take medication at a facility with a multispecialty team of doctors and medical experts who are skilled in CF diagnosis and treatment.

Medications

- Medications that target gene mutations, such as a new drug that combines three treatments to address the most prevalent genetic mutation that causes CF, which is seen as a big breakthrough in treatment.
- Antibiotics are used to treat and prevent infections in the lungs.
- Anti-inflammatory drugs can help reduce swelling in your lungs' airways.
- Mucus-thinning medications, such as hypertonic saline, can aid in coughing up mucus and enhance lung function.
- Bronchodilators are inhaled drugs that assist keep your airways open by relaxing the muscles surrounding your bronchial passages.
- Oral pancreatic enzymes to aid nutrient absorption in the digestive tract
- To avoid constipation or bowel obstruction, use stool softeners.
- Acid-reducing drugs can help pancreatic enzymes perform more efficiently.
- When diabetes or liver disease is present, specific medicines are prescribed.