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Pancreatic Disorders & Therapy

Perspective

Cystic Fibrosis: Diagnosis and Treatment

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DESCRIPTION

Cystic fibrosis (CF) is a disorder that damages your lungs, digestive tract and other organs. It's an inherited disease which is caused by a non-functioning gene. Cystic fibrosis affects the cells that produce mucus, sweat and digestive fluid. Mostly, these fluids are thin and smooth like olive oil. They lubricate your organs and tissues, keeping them from getting too dry. However, this genetic mutation increases the thickness of mucus and other body fluids. When this happens, function of organs is abnormally affected where the fluids become thicker.

Complications associated with Cystic fibrosis at more advanced cases, include respiratory failure and malnutrition.

Symptoms

People with CF can have symptoms including-

- Trouble with bowel movements
- Cough or trouble during breathing
- Frequent lung infections
- Trouble in gaining weight

Causes

Cystic fibrosis is genetic. People who have CF inherit two defective genes, one from each parent. Cystic Fibrosis (CF) is said to be recessive because you need to have two gene variants to have the condition itself. (An older name for gene variant is gene mutation.)

Your parents don't have to have cystic fibrosis for you to have CF. In fact, numerous families don't have a family history of Cystic Fibrosis (CF). If your family doesn't have a history of cystic fibrosis, the person with the gene variant is called the carrier. About 1 in 31 people in the U.S. is a carrier who is free of cystic fibrosis symptoms.

Diagnosis

Early diagnosis might be beneficial supporting in better healthier life. One among the mentioned tests can be used as diagnostic tool for the detection of cystic fibrosis

Blood test: The levels of Immuno Reactive Trypsinogen (IRT) is measured. Higher levels of IRT in patient's blood imply the presence of cystic fibrosis.

DNA test: Mutations to the CFTR gene are tested.

Sweat test: It measures the swab in sweat. Higher than normal results suggest CF.

Generally, the diagnosis test for cystic fibrosis (CF) is carried at the very earlier stage i.e, after birth itself. In any case, if this does not happen, such cases can be diagnosed only after certain period i.e at adult stage. Physicians might prescribe DNA or sweat test in case of presence of symptoms of the disease.

Treatment

Antibiotics like (Amoxicillin and clavulanic acid, Dicloxacillin) can prevent in treating lung infections and help the lungs work stronger. Administration of anti-inflammatory drugs such as ibuprofen and corticosteroids would be beneficial. Bronchodilators like hydrochloride (Xopenex®), albuterol (a metered dose inhaler), would help to dilation or relaxation of airways.

Consequently, utilization of Mucus thinners like (Hypertonic Saline, Mannitol (Bronchitol®) can help to get the soot out of your airways. Additionally, CFTR modulators can make your lungs work stronger. However it would increase weight. Else, use of Combination therapy can be immensely useful. The combination therapy including the drugs like elexacaftor/ivacaftor/tezacaftor (Trikafta) targets CFTR protein, resulting to make the drug work more efficiently.

Digestive problems

Enzymes produced by the pancreas to the small intestine aid in

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the process of digestion. However, mucus deposition might be a serious issue in digestion process. The absorption of digestive enzymes and required nutrients from the food cannot occur, which affects the following:

- Constipation
- Nausea, vomiting
- Swelling of abdomen
- Loss of appetite or reduced appetite
- Stunted growth in children

CONCLUSION

Cystic fibrosis is a systemic illness that has broad implications for both quality and quantity of the life, when inadequately controlled. Therefore, treatment should concentrate on optimizing mucus secretion in order to avoid acute illness events. Treatments should target to maintain lung function by controlling respiratory infection and clearing airways of mucus, optimizing nutritional status with pancreatic enzyme supplements and multivitamins, and eventually, by managing any other health complications that may arise.