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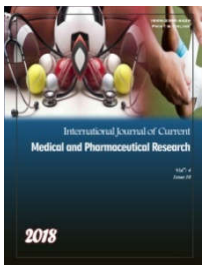

CYSTIC FIBROSIS

Hemavathy V., Girija Bhaskaran and Siyamaladevi M

Sree Balaji College of Nursing, No.7, CLC works road, Chrompet, Chennai-6000044

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ABSTRACT

Cystic fibrosis is a genetic disorder that affects mostly the lungs, but also the pancreas, liver, kidneys, and intestine. Long-term issues include difficulty breathing and coughing up mucus as a result of frequent lung infections. Other signs and symptoms may include sinus infections, poor growth, fatty stool, clubbing of the fingers and toes, and infertility in most males. Different people may have different degrees of symptoms.

INTRODUCTION

Definition

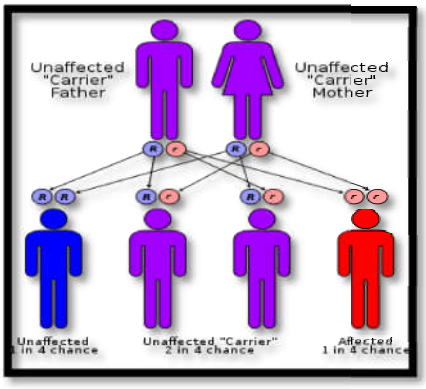
Cystic fibrosis is a hereditary disorder affecting the exocrine glands. It causes the production of abnormally thick mucus, leading to the blockage of the pancreatic ducts, intestines, and bronchi and often resulting in respiratory infection.

Risk groups

- Caucasians of Northern European descent.
- People who have a family history of cystic fibrosis

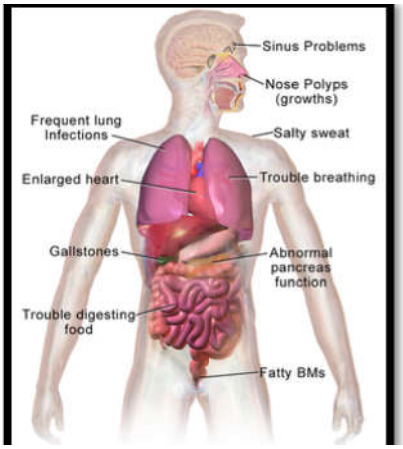
Etiology

CF is caused by a mutation in the gene cystic fibrosis transmembrane conductance regulator



Signs and symptoms

The symptoms of cystic fibrosis can vary depending on the person and the severity of the condition. The age at which symptoms develop can also differ. Symptoms may appear at infancy, but for other children, symptoms may not begin until after puberty or even later in life. As time passes, the symptoms associated with the disease may get better or worse.



One of the first signs of cystic fibrosis is a strong salty taste to the skin. Parents of children with cystic fibrosis have mentioned tasting this saltiness when kissing their children.

Other symptoms of cystic fibrosis result from complications that affect:

Corresponding author:* **Hemavathy V
Sree Balaji College of Nursing, No.7, CLC works road, Chrompet, Chennai-6000044,

- the lungs
- the pancreas
- the liver
- other glandular organs

Respiratory Problems

The thick, sticky mucus associated with cystic fibrosis often blocks the passageways that carry air into and out of the lungs. This can cause the following symptoms:

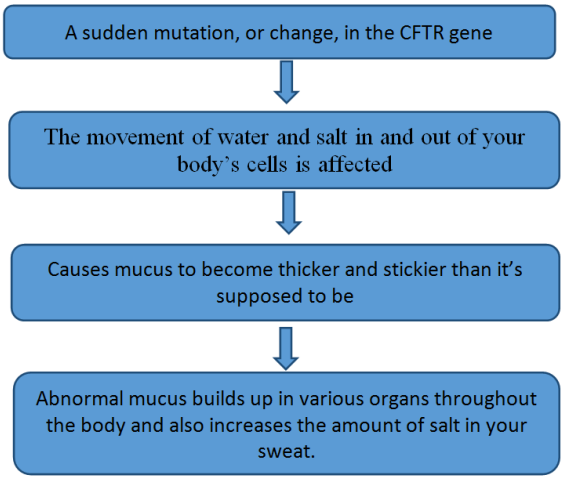
- wheezing
- a persistent cough that produces thick mucus or phlegm
- shortness of breath, especially when exercising
- recurrent lung infections
- a stuffy nose
- stuffy sinuses

Digestive Problems

The abnormal mucus can also plug up the channels that carry the enzymes produced by the pancreas to the small intestine. Without these digestive enzymes, the intestine can't absorb the necessary nutrients from food. This can result in:

- greasy, foul-smelling stools(steatorrhea)
- constipation
- nausea
- a swollen abdomen
- loss of appetite
- poor weight gain in children
- delayed growth in children

Pathophysiology



Diagnostic Evaluation

Immunoreactive Trypsinogen (IRT) Test:

The immunoreactive trypsinogen (IRT) test is a standard newborn screening test that checks for abnormal levels of the protein called IRT in the blood. A high level of IRT may be a sign of cystic fibrosis. However, further testing is required to confirm the diagnosis.

Sweat Chloride Test

The sweat chloride test is the most commonly used test for diagnosing cystic fibrosis. It checks for increased levels of salt in the sweat. The test is performed by using a chemical that makes the skin sweat when triggered by a weak electric current. Sweat is collected on a pad or paper and then analyzed. A diagnosis of cystic fibrosis is made if the sweat is saltier than normal.

Sputum Test

During a sputum test, the doctor takes a sample of mucus. The sample can confirm the presence of a lung infection. It can also show the types of germs that are present and determine which antibiotics work best to treat them.

Chest X-Ray

A chest X-ray is useful in revealing swelling in the lungs due to blockages in the respiratory passageways.
CT scan:

A CT scan creates detailed images of the body by using a combination of X-rays taken from many different directions. These images allows your doctor to view internal structures, such as the liver and pancreas, making it easier to assess the extent of organ damage caused by cystic fibrosis.

Pulmonary Function Tests (Pfts)

Pulmonary function tests (PFTs) determine whether your lungs are working properly. The tests can help measure how much air can be inhaled or exhaled and how well the lungs transport oxygen to the rest of the body. Any abnormalities in these functions may indicate cystic fibrosis.

Management

Although there's no cure for cystic fibrosis, there are various treatments available that may help relieve symptoms and reduce the risk of complications.

Medications

- Antibiotics may be prescribed to get rid of a lung infection and to prevent another infection from occurring in the future. They're usually given as liquids, tablets, or capsules. In more severe cases, injections or infusions of antibiotics can be given intravenously, or through a vein.
- Mucus-thinning medications make the mucus thinner and less sticky. They also help you to cough up the mucus so it leaves the lungs. This significantly improves lung function.
- Nonsteroidal anti-inflammatory drugs (NSAIDs), such as ibuprofen and indomethacin, may help reduce any pain and fever associated with cystic fibrosis.
- Bronchodilators relax the muscles around the tubes that carry air to the lungs, which helps increase airflow. You can take this medication through an inhaler or a nebulizer.
- Bowel surgery is an emergency surgery that involves the removal of a section of the bowel. It may be performed to relieve a blockage in the bowels.
- Cystic fibrosis may interfere with digestion and prevent the absorption of nutrients from food. A feeding tube to supply nutrition can be passed through the nose or surgically inserted directly into the stomach.
- A lung transplant involves removing a damaged lung and replacing it with a healthy one, usually from a deceased donor. The surgery may be necessary when someone with cystic fibrosis has severe breathing problems. In some cases, both lungs may need to be

replaced. This can potentially lead to serious complications after surgery, including pneumonia.

Surgical Procedures

- Lung transplantation often becomes necessary for individuals with CF as lung function and exercise tolerance decline. Although single lung transplantation is possible in other diseases, individuals with CF must have both lungs replaced because the remaining lung might contain bacteria that could infect the transplanted lung.
- A pancreatic or liver transplant may be performed at the same time to alleviate liver disease and/or diabetes.

Home Care

Cystic fibrosis can prevent the intestines from absorbing necessary nutrients from food. If you have cystic fibrosis, you might need up to 50 percent more calories per day than people who don't the disease. You may also need to take pancreatic enzyme capsules with every meal. Your doctor may also recommend antacids, multivitamins, and a diet high in fiber and salt.

If you have cystic fibrosis, you should do the following

- Drink plenty of fluids because they can help thin the mucus in the lungs.
- Exercise regularly to help loosen mucus in the airways. Walking, biking, and swimming are great options.
- Avoid smoke, pollen, and mold whenever possible. These irritants can make symptoms worse.
- Get influenza and pneumonia vaccinations regularly

Gene therapy

Gene therapy has been explored as a potential cure for CF. Results from clinical trials have shown limited success as of 2016, and using gene therapy as routine therapy is not suggested. The focus of much CF gene therapy research is aimed at trying to place a normal copy of the CTFR gene into affected cells. Transferring the normal CTFR gene into the affected epithelium cells would result in the production of functional CFTR protein in all target cells, without adverse reactions or an inflammation response. To prevent the lung manifestations of CF, only 5–10% the normal amount of CFTR gene expression is needed.

Phage therapy

Phage therapy is being studied for multidrug resistant bacteria in people with Cystic fibrosis.

Small molecules

A number of small molecules that aim at compensating various mutations of the CFTR gene are under development. One approach is to develop drugs that get the ribosome to overcome the stop codon and synthesize a full-length CFTR protein. About 10% of CF results from a premature stop codon in the DNA, leading to early termination of protein synthesis and truncated proteins. These drugs target nonsense mutations such as G542X, which consists of the amino acid glycine in position 542 being replaced by a stop codon. Aminoglycoside antibiotics interfere with protein synthesis and error-correction. In some cases, they can cause the cell to overcome a premature stop codon by inserting a random amino acid, thereby allowing expression of a full-length protein. The aminoglycoside gentamicin has been used to treat lung cells from CF patients in the laboratory to induce the cells to grow full-length proteins. Another drug targeting nonsense mutations is ataluren, which is undergoing clinical trials.

Prognosis

The outlook for people with cystic fibrosis has improved dramatically in recent years, largely due to advances in treatment. Today, many people with the disease live into their 40s and 50s, and even longer in some cases. However, there's no cure for cystic fibrosis, so lung function will steadily decline over time. The resulting damage to the lungs can cause severe breathing problems and other complications.

Prevention

Cystic fibrosis can't be prevented. However, genetic testing should be performed for couples who have cystic fibrosis or who have relatives with the disease. Genetic testing can determine a child's risk for cystic fibrosis by testing samples of blood or saliva from each parent. Tests can also be performed on you if you're pregnant and concerned about your baby's risk. Women who are pregnant or couples planning a pregnancy can have themselves tested for the CTFR gene mutations to determine the risk that their child will be born with CF. Testing is typically performed first on one or both parents and, if the risk of CF is high, testing on the fetus is performed

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