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Gene Technology

Commentary

Genetic Disorder: Cystic Fibrosis

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DESCRIPTION

Cystic Fibrosis (CF) is a genetic disorder that causes serious damage to the lungs, digestive system, and other body organs.

Cystic fibrosis affects cells that produce mucus, sweat, and digestive juices. These secreted fluids are usually thin and slippery. However, in people with cystic fibrosis, a defective gene causes the discharge to be sticky and thick. Instead of acting as a lubricant, secretions block ducts, ducts, and passageways, especially in the lungs and pancreas.

The signs and symptoms of cystic fibrosis depend on the severity of the disease. Even in the same person, symptoms may worsen or improve over time. Some people may not experience symptoms until adolescence or adulthood. People not diagnosed before adulthood usually have a milder course of the disease and are more prone to atypical symptoms such as recurrent attacks of inflammation of the pancreas (pancreatitis), infertility, and recurrent pneumonia.

People with cystic fibrosis have higher than normal levels of salt in their sweat. Many of the other signs and symptoms of cystic fibrosis affect the respiratory and digestive systems.

Respiratory signs and symptoms

Thick, sticky mucus associated with cystic fibrosis blocks the tubes that carry air into and out of the lungs. This can cause the following signs and symptoms:

- Persistent cough with thick mucus (sputum)
- Wheezing
- Exercise intolerance
- Recurrent lung infection
- Nasal inflammation or congestion
- Recurrent sinusitis

Gastrointestinal signs and symptoms

Thick mucus can also block the ducts that carry digestive enzymes from the pancreas to the small intestine. Without these digestive enzymes, your intestines cannot fully absorb the nutrients from the food you eat. The result is often:

- Fatty, foul-smelling stools
- Weight gain and poor growth
- Intestinal obstruction (meconium ileus), especially in new bourns
- Chronic or severe constipation, which may include frequent straining when attempting to defecate, may eventually lead to partial rectum protrusion. anus (rectal prolapse)

In cystic fibrosis, a defect (mutation) in the Cystic Fibrosis Transmembrane Conduction Regulator (CFTR) gene changes the protein that controls the movement of salts into and out of cells. The result is sticky mucus in the respiratory, digestive, and reproductive systems, and increased salt content in sweat.

Gene can have a variety of defects. The type of gene mutation is related to the severity of the condition.

A child must inherit one copy of a gene from each parent to develop the disease. If children inherit only one copy, they won`t develop cystic fibrosis.

Respiratory complications

Respiratory tract involvement (bronchiectasis): Cystic fibrosis is one of the leading causes of bronchiectasis, a chronic lung disease characterized by abnormal enlargement and scarring of the airways (bronchial tubes). This makes it difficult for air to enter and exit the lungs and makes it difficult to clear mucus from the bronchi.

Chronic infection: The thick mucus in the lungs and sinuses is an ideal breeding ground for bacteria and fungi. People with cystic fibrosis often have a sinus infection, bronchitis, or pneumonia. Infections are common with bacteria that are resistant to antibiotics and difficult to treat.

Nasal growth (nasal polyps): Soft, fleshy growths (polyps) can develop because the lining inside the nose becomes inflamed and swollen.

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Blood vomiting (hemoptysis): Bronchiectasis may develop near blood vessels in the lungs. The combination of airway damage and infection can make you vomit blood. Often this is a small amount of blood, but it can also be life-threatening.

Pneumothorax: In this condition, air leaks into the space separating the chest wall and lungs, causing the lungs to partially or completely collapse. It is more common in adults with cystic fibrosis. Pneumothorax can cause sudden chest pain and shortness of breath. People often feel their chest simmering.

Respiratory failure: Over time, cystic fibrosis can severely damage lung tissue and stop working. Lung function usually worsens gradually, and it eventually can become life threatening. Respiratory failure is the most common cause of death.

Acute exacerbations: People with cystic fibrosis may experience worsening of their respiratory symptoms, such as coughing with more mucus and shortness of breath. This is called an acute exacerbation and requires treatment with antibiotics. Sometimes treatment can be provided at home, but hospitalization may be needed. Decreased energy and weight loss also are common during exacerbations.

Digestive system complications

Liver disease: The tube that carries bile from your liver and gallbladder to your small intestine may become blocked and inflamed. This can lead to liver problems, such as jaundice, fatty liver disease and cirrhosis and sometimes gallstones.

Nutritional deficiencies: Thick mucus can block the tubes that carry digestive enzymes from your pancreas to your intestines. Without these enzymes, your body cannot absorb protein, fat, or fat-soluble vitamins, so you cannot get enough nutrients. This can lead to stunted growth, weight loss, or inflammation of the pancreas.

Diabetes: The pancreas produces the insulin the body needs to use sugar. Cystic fibrosis increases the risk of diabetes. Diabetes occurs in about 20% of adolescents with cystic fibrosis and in 40-50% of adults.

Other complications

Electrolyte imbalances and dehydration: Because people with cystic fibrosis have saltier sweat, the balance of minerals in their blood may be upset. This makes it easy to become dehydrated, especially when exercising or in hot weather. Signs and symptoms include increased heart rate, fatigue, weakness, and low blood pressure.

Mental health problem: Treating chronic conditions that do not respond to treatment can lead to fear, depression, and anxiety.