Health Basics

Cystic Fibrosis

What is cystic fibrosis?

Cystic fibrosis (CF) is a progressive genetic disease that severely affects the ability of the lungs, pancreas and other organs to function properly, according to the Cystic Fibrosis Foundation.

A defective gene causes thick, sticky mucus buildup. The mucus clogs the lungs' airways and traps bacteria, causing infections, extensive lung damage and respiratory failure. Because of the mucus, the pancreas is unable to release digestive enzymes that allow the body to break down food and absorb important nutrients.

More than 30,000 people are living with cystic fibrosis in the United States, and 1,000 new cases are diagnosed each year. In addition, more than 75 percent of people with CF are diagnosed by age 2, and more than half of the CF population is age 18 and older.

What are the signs and symptoms of cystic fibrosis?

Symptoms associated with CF, include:

- Very salty-tasting skin
- Persistent coughing, at times with phlegm
- Frequent lung infections, including pneumonia or bronchitis
- Wheezing or shortness of breath
- Poor growth or weight gain in spite of a good appetite
- Frequent greasy, bulky stools or difficulty with bowel movements
- Infertility in most males and reduced fertility in females.

CF can lead to complications in the respiratory, digestive and reproductive systems, such as damaged airways, respiratory failure, diabetes, nutritional deficiencies, infertility or being less fertile and much more.

What are the risk factors for cystic fibrosis?

- Family history: Cystic fibrosis is an inherited disorder, which means it runs in families. To develop CF, children must inherit one copy of a defective gene from each parent. Those with only one copy won't acquire CF, but they become carriers who can potentially pass the gene to their own children.
- Race: The disease can occur in all races, but it is most commonly found in white people of Northern European ancestry.

How is cystic fibrosis diagnosed?

Every state in the United States routinely screens newborns for CF in an effort to treat it early if diagnosed.

These screenings include a test that assesses blood samples for high levels of immunoreactive trypsinogen (a chemical released by the pancreas), genetic exams to confirm diagnosis and to check for specific defects on the gene responsible for CF, and sweat tests to determine whether the skin is saltier than normal.

In addition, diagnostic exams are recommended for older children and adults who weren't screened at birth.

How is cystic fibrosis treated?

Although cystic fibrosis is an incurable disease, different treatments may help ease symptoms and reduce complications.

Doctors prescribe antibiotics to treat and prevent lung infections, anti-inflammatory meds to lessen swelling in the airways in the lungs and mucus-thinning drugs to help patients cough up the thick secretions, which could improve lung function. In addition, inhaled medications and oral pancreatic enzymes are also used to keep airways open.

Chest physical therapy also helps to loosen mucus in the lungs. Common techniques include using cupped hands to clap on the front and back of the chest and special breathing methods. Additionally, doctors may recommend that patients use mechanical devices, such as vibrating vests or breathing tubes and masks.

Pulmonary rehabilitation, a long-term program of exercise and breathing techniques, along with education about CF, advice about nutrition, counseling and support may also improve lung function and overall well-being. Doctors may also turn to procedures such as nasal polyp removal, oxygen therapy and lung transplantation to help improve breathing and blood oxygen levels.

How can cystic fibrosis be prevented?

If any close relatives have CF, partners may want to undergo genetic screening before having

children. Such testing can help determine the risk of having a child with the disease.

For women who are already pregnant with a baby at risk for CF, further tests can be conducted on the developing child.

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