

Understanding Cystic Fibrosis

Over 30,000 people in America are living with cystic fibrosis.

1 Introduction

Cystic fibrosis (CF) is a genetic disease that occurs due to a mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. This gene codes for a protein that, in healthy individuals, helps to keep the mucus that lines organ membranes thin and watery. In patients with CF, the proteins are translated incorrectly and, as a result, the mucus thickens and builds up. This buildup is especially dangerous in the lungs as it obstructs airways and traps harmful bacteria. CF is a chronic and progressive disease, and, as of now, there is no cure.

2 Signs and Symptoms

CF is typically detected at or before childhood, with diagnoses occurring in as early as newborns. Some of the key symptoms that may indicate that a child may have CF are:

- Decreased rate of weight gain at birth
- Salty-tasting skin
- Loose or oily stools
- Trouble breathing or wheezing
- Recurrent sinus infections
- Frequent coughing

3 Risk Factors

In order for cystic fibrosis to occur, two faulty copies of the CFTR gene must be present. This requires for both parents to have at least one copy of the defective gene. If only one parent has the gene, their child will either become a carrier of CF or be unaffected. However, if both parents have at least one copy of the mutated gene, the child will have a:

- 25% chance of having CF
- 50% chance of being a carrier
- 25% chance of being neither a carrier nor having CF

4 Treatments

Although there is no cure for CF, there are treatments and medications that can help alleviate symptoms. Many medications are focused on improving lung function, such as mucus thinners and bronchodilators. Others aim to remedy the intestinal and digestive issues that result from the mucus buildups.

There are also relatively new treatments that target the faulty proteins that are made by the mutated CFTR gene. These drugs help to improve the function of the proteins, therefore decreasing the downstream effects of CF.

Did you know?

There are more than 1,700 known mutations of the CFTR gene.

For more information on cystic fibrosis and supportive resources please visit www.cff.org.

References:

<https://www.mayoclinic.org/diseases-conditions/cystic-fibrosis/diagnosis-treatment/drc-20353706>

<https://my.clevelandclinic.org/health/diseases/9358-cystic-fibrosis>