*OTHERS:*

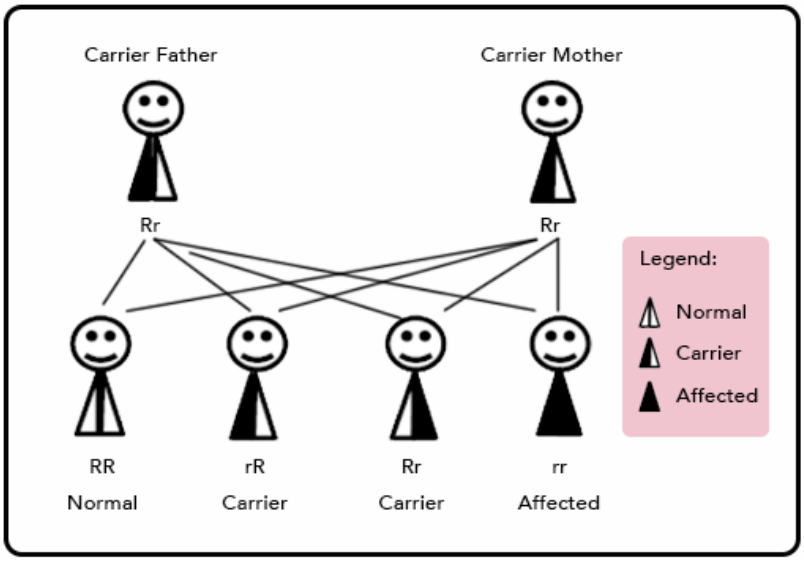
# CYSTIC FIBROSIS

## What is Cystic Fibrosis?

Cystic Fibrosis (CF) is a progressive, inherited condition caused by build-up of sticky and thick mucus that can damage the body's organs. Untreated children born with this condition may have serious chronic health effects that could lead to early death. Fortunately, this disorder can be detected early through newborn screening. Early detection prevents the complications of this condition.

## What causes Cystic Fibrosis?

Cystic Fibrosis occurs when a particular cell protein called **"cystic fibrosis transmembrane conductance regulator" (CFTR)** is either missing or not working well. The **CFTR gene** provides instructions for making a channel that transports chloride ions into and out of the cells. Mutations in the CFTR gene cause disruptions in the flow of chloride ions and water across cell membranes. Without the chloride to attract water to the cell surface, the mucus in various organs becomes thick and sticky. This may involve the cells that line passageways of the lungs and the digestive system including the pancreas, liver, and gallbladder.




Cystic Fibrosis is an **autosomal recessive** inherited condition. It affects both boys and girls equally. Everyone has a pair of genes that make the **CFTR protein**. **In children with CF, both of these genes are not working correctly.** These children inherit one non-working gene for the condition from each parent. On the other hand, **parents of children with CF usually do not have the disorder**. They only have a single non-working gene for CF and are called **carriers**. Carriers do not have CF because the other gene of this pair is working correctly. When both parents are carriers, for every pregnancy there is a 25% chance for the child to have CF, 50% chance for the child to be a carrier just like the parents and 25% chance for the child to have two working genes.

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## WHAT ARE THE SIGNS AND SYMPTOMS OF CYSTIC FIBROSIS?

Clinical manifestations usually start in early childhood. Most children with CF show signs and symptoms before one year of age. There are also some people who do not find out they have CF until adulthood.

Since CF is a **multi-organ, clinically diverse disorder**, awareness of pulmonary and extrapulmonary manifestations are important in providing appropriate care for these patients. Children born with this condition may have **salty sweat** (many parents notice a salty taste when kissing their child). Accumulation of thick mucus and phlegm **results in constant coughing, frequent lung and sinus infections** (pneumonias and bronchitis), shortness of breath, nasal polyps (fleshy growths inside the nose), and eventually permanent lung damage. Digestive problems include **meconium ileus** (bowel obstruction that occurs when the meconium in your child's intestine is very thick and sticky creating a blockage in a part of the small intestine called the ileus), **poor weight gain and growth** (even when a baby or child eats a lot) greasy and smelly stools (that are also bulky and pale colored), intestinal problems (diarrhea or constipation, abdominal pain or gassiness), and rectal prolapse (protrusion of the rectum through the anus). Liver disease, diabetes, pancreatitis, gallstones, and even male infertility may also occur in some patients with CF.

## WHAT IS THE TREATMENT OF CYSTIC FIBROSIS?

Cystic Fibrosis is a complex disease and the types and severity of symptoms can differ widely from person to person. **The main goal of treatment is to keep the lungs clear of thick mucus and to provide the correct amount of calories and nutrients to keep the patient healthy.** This would include supplements (vitamins A, D, E, and K), pancreatic enzyme supplement, a well-balanced, high-caloric diet that is low in fat and high in protein, CFTR modulators, and an individualized fitness plan to help improve energy, lung function and overall health. Medications such as laxatives and mucolytic agents may also be used to treat intestinal and airway obstructions.

## REFERENCES:


Cystic Fibrosis Foundation. www.cff.org. Accessed June 28, 2021.
Cystic Fibrosis Fact Sheet.www.cdc.gov. Accessed June 29, 2021.
Baby's first test. https://www.babysfirsttest.org/newborn-screening/conditions/cystic-fibrosis-cf. Accessed June 29, 2021.


## OTHER IMPORTANT CONSIDERATIONS

- Children with CF should be given all the usual childhood vaccinations according to the regular schedule. Especially important are the vaccines against measles, influenza, and pneumonia.
- They should be kept away from smoke exposure, especially from cigarette smoke as this can add to lung damage.
- Frequent hand washing is encouraged for patients and caregivers to prevent the spread of infection.
- Incorporate regular physical activity and exercise into daily activities to maintain the child's lung function and improve overall health.
- Allow the child to drink more water and fluids to help loosen the thick mucus and to prevent dehydration. Children with CF lose more salt than others, especially during exercise or in hot weather.
- It is best to consult with your doctor immediately if the child has a respiratory infection and is too sick to eat or follow regular health habits.