

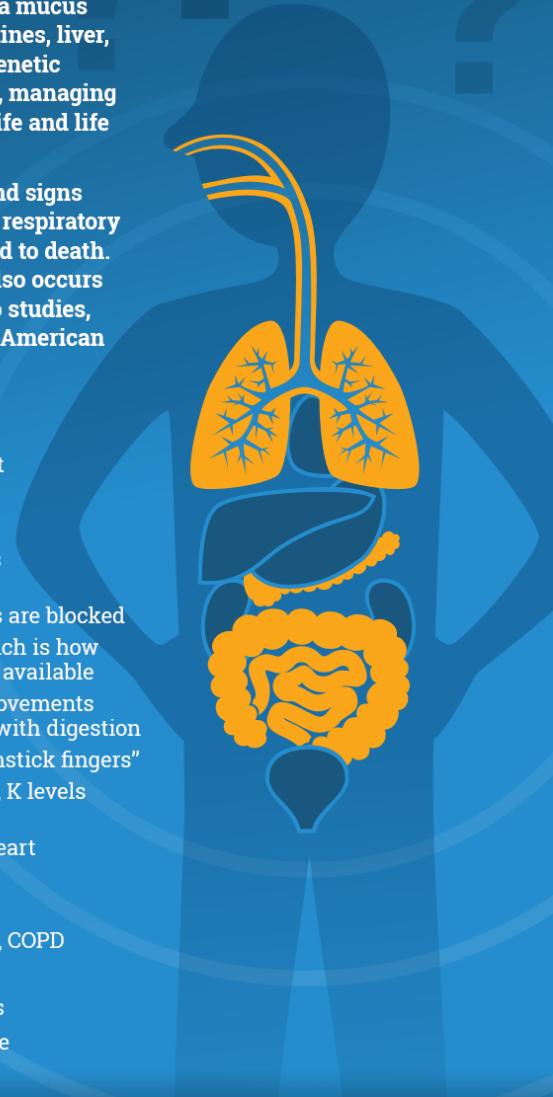
# What is CYSTIC FIBROSIS?

Cystic Fibrosis causes the cells of the body to produce mucus that is thick and sticky. Inability to clear this creates a mucus build-up affecting the lungs, sinuses, pancreas, intestines, liver, and reproductive organs. Cystic Fibrosis is a severe genetic disease but with the advancement in medical science, managing the disease through treatment to increase quality of life and life expectancy is possible.

There is no cure for Cystic Fibrosis. The symptoms and signs vary, but it is commonly diagnosed in children due to respiratory symptoms and digestive complications which can lead to death. It is most common in the caucasian population, but also occurs in less frequency in other ethnic groups. According to studies, the disease occurs in 1 out of 2,500 - 3,500 Caucasian-American newborns in the United States.

## Symptoms include:

- Coughing and lung infections that are difficult to treat
- Constant thick mucus in the lungs and sinuses
- Recurring sinus infections
- Weight loss and failure to thrive (being small for one's age and not growing as expected)
- Meconium ileus: a condition where a baby's intestines are blocked
- People with Cystic Fibrosis have very salty sweat, which is how the disease was diagnosed before genetic testing was available
- Frequent bathroom visits with foul-smelling bowel movements and pain after eating fatty foods; and other problems with digestion
- Finger clubbing: enlarged fingertips, also called "drumstick fingers"
- Problems relating to malnutrition: low vitamin A, D, E, K levels (these are the fat-soluble vitamins) and low iron
- Problems with other organs such as pancreas, liver, heart and spleen in addition to the typical symptoms
- Typical in teenage or adult patients:
  - Secondary lung complications like bronchiectasis, COPD
  - Cystic Fibrosis related diabetes
  - Low bone density which could lead to osteoporosis
  - Reduction in reproductive function, including male infertility due to an absent vas deferens

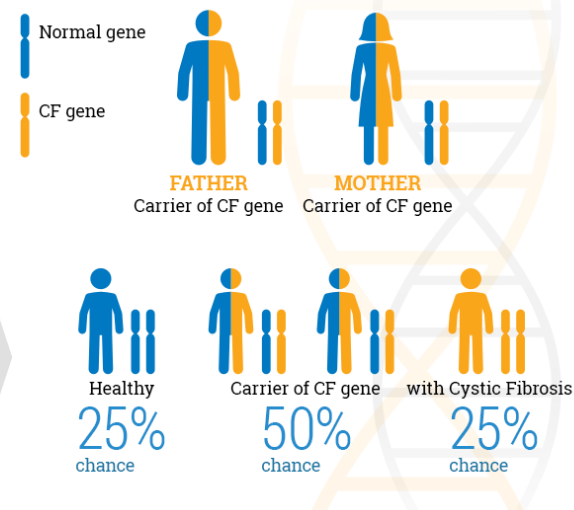


## A GENETIC DISEASE

Cystic Fibrosis is a genetic disease caused by mutations in a gene. Cystic Fibrosis is an autosomal recessive disease, which means a person will have the disease if he inherits two copies of the mutated gene.

There are more than 2000 mutations in the CFTR gene already identified, however the most common mutation is DeltaF508.

Parents of children with Cystic Fibrosis are carriers, meaning they carry one copy of the mutated gene. Carriers do not show any symptoms of the disease, but they can pass their mutated gene to their children. With two parent carriers, the risk to each child of having Cystic Fibrosis is as follows:



# CFTR

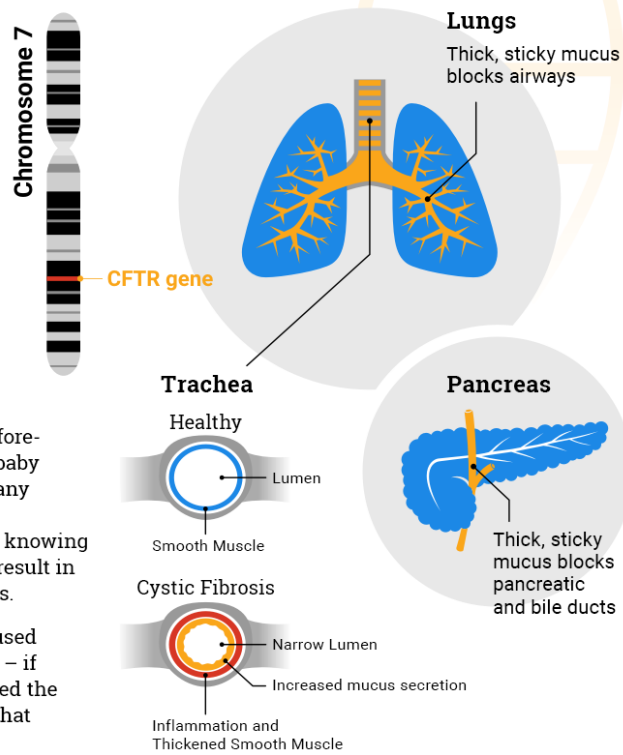
The Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) is a membrane protein and chloride channel that receives instruction from the CFTR gene, to transport chloride ions in and out of cells. Chloride ions play a key role in controlling the movement of water in tissues. In Cystic Fibrosis, the chloride ions are unable to properly regulate the flow which results in abnormal mucus.

## Diagnosis

**Genetic testing** is considered very essential. Through genetic testing, parents can learn beforehand if they are carriers of Cystic Fibrosis. A baby can be diagnosed with Cystic Fibrosis before any symptoms start appearing, which allows for preventative and prompt treatment. Moreover, knowing the specific mutations of the CFTR gene may result in treatment with new gene targeted medications.



A **sweat test** is also commonly used as a diagnostic test on a patient – if the sodium chloride levels exceed the normal ranges, it is very likely that the person has Cystic Fibrosis.



# TREATMENT

Over the past few decades advances in research and treatments have significantly improved the life expectancy of people with Cystic Fibrosis, however it remains a very serious life threatening condition. The outcome of the patient depends on many factors – some patients are living into their fifties, however some patients pass away as children.

It is always recommended to start the treatment as soon as possible to increase the quality and longevity of life. People with Cystic Fibrosis can live relatively normal lives, but they need to adhere to all their prescribed treatments, and often must make accommodations in school, work and other areas. Treatments and hospitalizations vary with the severity of the disease presentation and the stage of the disease, but often include:



- Medication to open the airways and reduce swelling of the airways (Bronchodilators and steroids usually inhaled through an inhaler pump or nebulizer)
- Respiratory therapies and airway clearance techniques several times a day: includes chest physiotherapy, breathing exercises, exercise, the Vest™ (a machine designed to help a patient do airway clearance), and other devices
- Medication to break down the thick mucus (Mucolytics)
- Antibiotics to treat infection: oral, inhaled through a nebulizer, or administered via an IV
- Enzyme tablets to assist with digestion of food, especially protein and fat
- Medications targeted at restoring the CFTR function
- A diet high in calories, fat and protein and nutritional supplements to help weight gain
- Prescribed vitamin supplements to combat low fat-soluble vitamin A, D, E, K levels
- Supplements and medication to improve bone density
- Medication to treat any secondary complications, on a case-by-case basis: e.g. insulin for diabetes
- Supplemental oxygen for low saturation (in advanced stages)
- Transplant (in advanced stages)

**It is important to receive treatment from a medical center that specializes in Cystic Fibrosis treatment, not just a regular pulmonologist.** It is possible for a person to go undiagnosed if their initial symptoms are mild, however, Cystic Fibrosis is a progressive disease so if the person doesn't receive care, their symptoms can worsen. If you suspect you or someone you know may have Cystic Fibrosis, insist on a Cystic Fibrosis sweat screening test and a genetic test performed at an accredited Cystic Fibrosis center.

This information is for educational purposes only. Refer to your doctor for medical advice.