What is Cystic Fibrosis?

Cystic Fibrosis or known as CF is an inherited genetic disease that causes buildup of a thick sticky mucus in the body. The mucus blocks vital passageways and ducts making it difficult for the body to work normally.

History of CF

CF has existed for centuries and can be referred through medical works as early as 1595.
In 1936, Swiss pediatrician Guida Fanconi named the disease. Later in 1938, American Pathologist Dr. Dorothy Anderson provided the first description of the disorder in medical literature. She discovered it during an autopsy of a child and called it "cystic fibrosis of the pancreas." Following the discovery, Dr.Anderson continued researching the diseases and found methods to diagnose it within living patients.

Dr.Anderson accepting an award for discovering Cystic Fibrosis.

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Cystic Fibrosis

A genetic disorder

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How is it inherited?

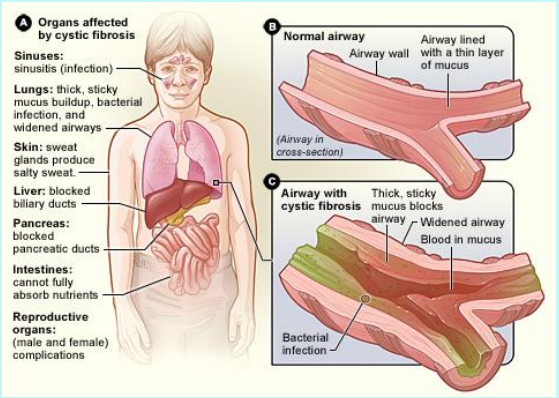
CF is autosomal recessive, and is caused by a mutation in chromosome 7 affecting the CFTR gene. CF is inherited when both CF parents pass down the CFTR gene mutation to the child.

Inheritance Punnett Square

Rr x Rr

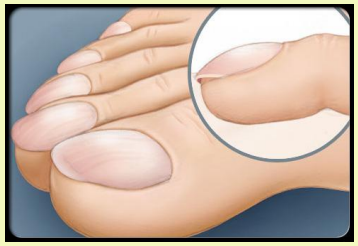
	R	r
R	RR	Rr
r	Rr	rr

RR= not affected
Rr= carrier
rr= has disease



Signs/Symptoms

- oral mucus discharge
- Frequent coughing
- Pneumonia
- Wheezing
- Salty sweat
- Fatigue
- bulky/light bowel movements
- Clubbing toes or fingers



CF Frequency

- 60,000 people have CF worldwide
- 30,000 people have CF in the U.S.
- 10–12 million people are carriers in the U.S.
- 1 out of every 3,500 newborn has CF
- CF mutations largely occur in specific ethnic groups, mainly European derived groups

Diagnosis

If a doctor believes a patient has CF, a sweat test is performed. If the test shows that the patient has a high level of chloride, the patient is likely to have CF. In some states newborns are blood tested for CF. Although, a blood test is not as accurate as a sweat test.

Infant receiving a sweat test

Treatment

Although CF has no cure, there are a few methods to help the body work as normal.

- Coughing is used to clear lung mucus
- Antibiotics help fight or prevent lung infections
- Chest physical therapy helps to drain mucus
- Therapy vests shake the chest allowing the patient to have independent therapy
- Enzymes can be taken through mouth to aid digestion for digestive CF