

# Genetics and Cystic Fibrosis: One Disease, Many Mutations

CHROMOSOME 7

The gene that causes CF, known as the **cystic fibrosis transmembrane regulator (CFTR) gene**, was discovered in 1989.

## THE FACTS



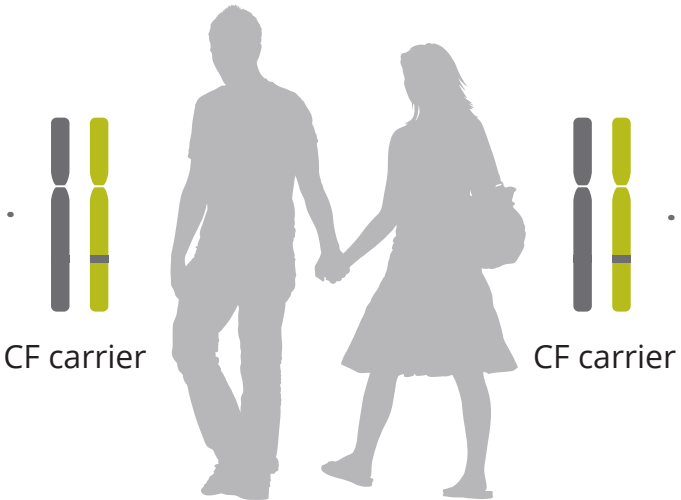
Since 1989, scientists have identified approximately **2,000 CFTR mutations.**

**127**

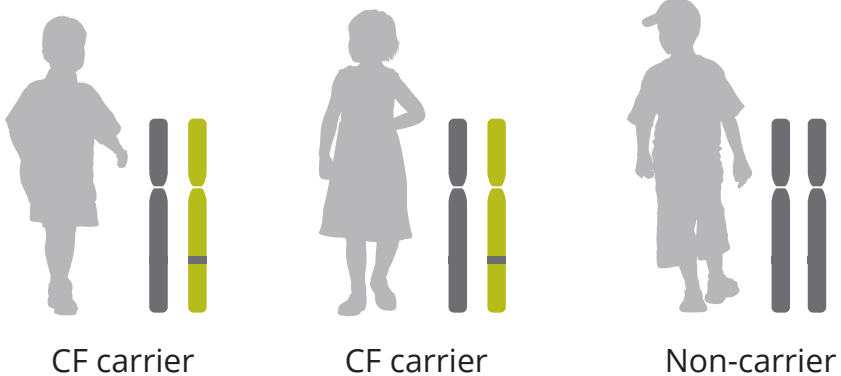
CFTR mutations are known to cause CF.

## genetics of cystic fibrosis

A CF carrier is a person who inherits a defective CFTR gene from one parent, and a normal copy from the other.



**1 in 4** the chance that a child will have CF when both parents are carriers of a defective CFTR gene



### Child with CF

A child must inherit **2 defective CFTR genes** – one from their mother, and one from their father – to have CF.

*CF is an autosomal recessive disorder – both copies of the gene in each cell must have mutations.*