The Genetics of Cystic Fibrosis

Cystic fibrosis (CF) is a genetic (inherited) disease. It is caused by a faulty gene that affects the body's cells, tissues, and glands that make mucus and sweat. A child will be born with CF only if they inherit 2 CF genes, 1 from each parent. A person who has only 1 CF gene is called a CF carrier. They are healthy and don't have the disease. But they are a carrier of the disease. A parent can be a CF carrier and pass the CF gene on to their child.

Once parents have had a child with CF, they have a 1 in 4 chance that each additional child will be born with CF. This means there is a 3 out of 4 chance that additional children won't have CF. But these children may be carriers of the CF gene.

Most often, a family has no history of CF. So the diagnosis comes as a surprise to parents. Both parents are healthy, so they didn't know that they carried the gene. Or that they passed the gene to the unborn baby at the same time.

Genes are found on structures in the cells of the body called chromosomes. Each cell normally has 46 total chromosomes or 23 pairs of chromosomes. The seventh pair of chromosomes has a gene called the CFTR (cystic fibrosis transmembrane conductance regulator) gene. Changes (mutations) or errors in this gene are what cause CF. This gene is very large and complex. More than 1,800 different mutations in this gene have been found that cause CF. The abnormal CFTR gene may result in no CFTR protein being made. Or it may lead to a defective CFTR protein that cannot work correctly in the cell.

According to the Cystic Fibrosis Foundation, if you have no family history of CF, the risk of having a mutation in the gene for CF depends on your ethnic background:

Ethnic background	Risk of CF mutation	Risk of child with CF
Caucasian American	1 in 29	1 in 2,500 to 3,500
Hispanic American	1 in 46	1 in 4,000 to 10,000
African American	1 in 65	1 in 15,000 to 20,000
Asian American	1 in 90	1 in 100,000

Screening for CF is part of newborn screening in every state in the U.S. A positive newborn screening is not a diagnosis of CF. But it does mean more testing is done.

The first test done to try to diagnose CF is a sweat chloride test. This measures the amount of salt in your child's sweat.

Testing for the CF gene can be done from a small blood sample. Or it can be done with a cheek swab. For this, a brush is rubbed against the inside of your cheek. This is done to get cells for testing. Labs generally test for the 20 or so most common mutations.

Not all of the genetic errors that cause CF have been found. Many people with CF have mutations that have not yet been identified. Not all mutations can be found. So you can still be a CF carrier even if no mutations were found by carrier testing.

Testing for the CF gene is advised for anyone who has a family member with the disease. It's also advised if your partner is a known carrier of CF. Or if they are affected by CF.