Adult CF: The Genetics of Cystic Fibrosis

Cystic fibrosis (CF) is a genetic disease. This means that CF is inherited. A person is born with CF only if two CF genes are inherited. You have to get one from your mother and the other from your father.

A person who has only one CF gene will not have the disease. But they are a carrier of the disease. If a CF-gene carrier has a child with another CF-gene carrier, they will have an increased chance of having a child with CF.

When two CF-gene carriers have a child, they have a 1 in 4 chance that the child will be born with CF. This means that there is a 3 in 4 chance that the child will not have CF. But this child may be a carrier of the CF gene.

Often both parents have no family history of CF. So a diagnosis may come as a surprise to them. Since both parents are healthy, they didn't know that each one carried the CF gene.

Genes are found on structures (chromosomes) in the cells of the body. Each cell normally contains 46 total chromosomes, or 23 pairs. The seventh pair of chromosomes contains a gene called the cystic fibrosis transmembrane conductance regulator (CFTR) gene. Changes (mutations) in this gene are what cause CF. This gene is quite large and complex. More than 1,700 different mutations in this gene have been found that cause CF.

People of northern European ancestry are more likely to have CF. It's less common in African Americans, Hispanics, and Asian Americans.

Diagnosis

In the U.S., CF is usually diagnosed in children by age 2, as newborns are now routinely screened for the disease. But such testing was not required in all U.S. states until 2010. As a result, CF is sometimes not diagnosed until late childhood or even adulthood. Testing for the CF gene is recommended for anyone who has a family member with the disease.

Testing for the CF gene can be done from a blood sample. Or it may be done from a cheek swab, which is a brush rubbed against the inside of your cheek to get cells for testing. Labs generally test for 20 or more common mutations.

Many people with CF have mutations that have not been identified. In other words, all of the genetic problems that cause the disease have not been found. Because not all mutations are detectable, a person can still have CF or be a CF-gene carrier even if no mutations were found.

If testing for CF comes back positive, you'll likely have a sweat chloride test. This test measures the amount of salt in your sweat. People with CF tend to have high levels of chloride, which is found in salt, in their sweat. A sweat chloride test is often the first test done to try to diagnose CF.