



Cystic Fibrosis Carrier Testing

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What is Cystic Fibrosis (CF)?

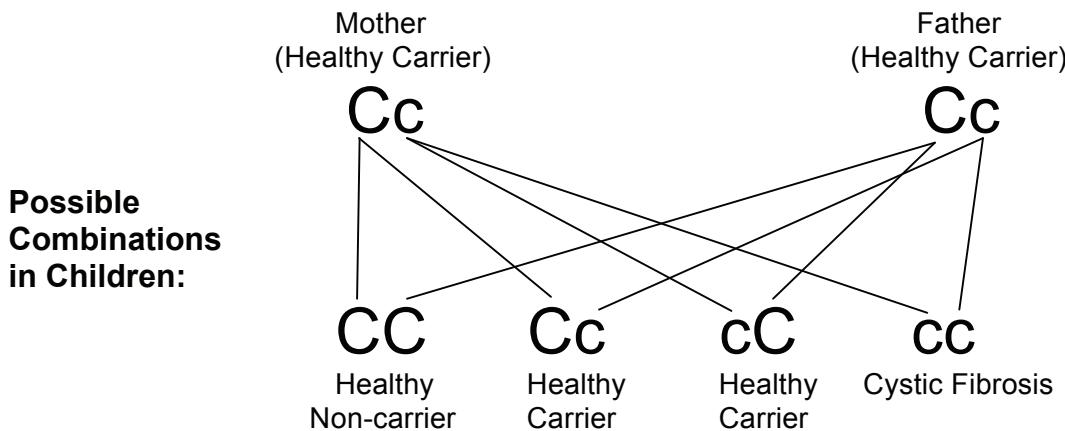
Classic cystic fibrosis is a genetic disease which causes changes in the secretions of the body. Thick mucus plugs the lungs and leads to chronic and severe respiratory problems. Pancreatic ducts are blocked and this can interfere with digestion. Individuals with classic CF have an average life span of 36.9 years, although some die much earlier. Death is primarily due to respiratory failure and infection. Classic CF often leads to repeated hospitalization throughout life. Intelligence is usually normal. In addition to classic CF, there are some variant types which have less severe symptoms including milder respiratory problems, sinusitis, or infertility.

How is Cystic Fibrosis inherited?

CF is an autosomal recessive genetic disorder caused by mutations in a particular gene (called the *CFTR* gene). Everyone has two copies of this gene. A person with one normal copy and one abnormal copy is a **carrier** of CF. Carriers usually do not have disease symptoms (there are some exceptions). A person will have CF **disease** if he/she inherits an abnormal copy of the *CFTR* gene from **both** of their parents.

Inheritance of Cystic Fibrosis:

[C = normal *CFTR* gene & c = *CFTR* gene with a mutation]



What is the risk of having a child with Cystic Fibrosis?

As diagrammed above, if both parents are carriers, there is a one in four risk with **each** pregnancy that their child will inherit both abnormal copies of the gene and have CF.

How common is Cystic Fibrosis and how many people are carriers?

The answer to that question depends on your racial/ethnic background. As you can see by the first three columns of the following table, CF is more common in Caucasians, Ashkenazi Jews, and some Native Americans. It is less common in Hispanics, African-Americans, and Asian Americans.

Group	Number of People with CF	Chance to be a CF Carrier	Accuracy of DNA Test
Caucasians	1/3,300	1 in 29 people	88%
Hispanics	1/8,000-9,000	1 in 46 people	72%
Ashkenazi Jews	1/3,300	1 in 29 people	94%
Native Americans	1/1,500- 1/3,970	unknown	94%
African Americans	1/15,300	1 in 60-65 people	65%
Asian Americans	1/32,100	1 in 90 people	49%

Is there a test to determine if I am a CF Carrier?

Genetic testing for CF is available. The test is done by studying the DNA from the cheek cells or from a blood sample. If your test detects an abnormal copy of the gene, then the test is said to be positive. If the test does not detect an abnormal copy, then the test is said to be negative. The last column in the previous table gives the proportions of mutations that can be found by this test. You will note that we cannot yet detect all mutations in the CF gene (see table on other side).

What does it mean if my CF carrier test is positive?

If the laboratory says your test is positive, that means they have identified a mutation in one of your copies of the CF gene. However, because everyone has two copies of the gene, your other normal copy keeps you from ever having CF. People with CF have a mutation in *both* copies. If you test positive, then your partner should have carrier testing. If both of you are found to be carriers, prenatal testing is offered for your pregnancies because you have a one in four (25%) chance with each pregnancy of having a child with CF. In order to be eligible for CF prenatal testing, both you and your partner must complete CF carrier testing by your 16th week of pregnancy, or no later than 2 weeks after you have an amniocentesis. Genetic counseling is recommended if your test is positive.

What does it mean if my CF carrier test is negative?

If the test is negative, it means the laboratory did not identify a mutation in either copy of your CF gene. Because this test is not 100% accurate (see table, column 4), there is a small chance that you still carry a rare mutation the test missed. That is, you could still be a carrier. Prenatal testing is definitive when the laboratory is able to identify a CF mutation in both you and your partner.

Is carrier testing recommended?

Carrier testing is recommended for anyone who has a family member with cystic fibrosis or anyone who has a partner with the disease. In addition, carrier testing for CF is recommended for non-Jewish Caucasians and Ashkenazi Jewish individuals by the American College of Medical Genetics (ACMG) and the American College of Obstetrics & Gynecology (ACOG). Screening is available for other ethnic groups; however, the chance of being a carrier is lower, as is the detection rate (see table on other side).