Laboratories and Genetic Service Centers

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7315 SW 87th Avenue
Miami, FL 33173
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Signal Hill, CA 90806
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3 Garret Mountain Plaza, 3rd Floor
West Paterson, NJ 07424
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107 Scripps Drive, Suite 110
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Winter Park, FL 32789
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4600 Habana Avenue, N Suite 12
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INTEGRATED GENETICS

Cystic Fibrosis DNA Testing

What is the CF Carrier Test?

There is now a test which can detect the most common disease-causing changes in the CF gene, and which therefore can find most of the people who are CF carriers. The test can be done on a small sample of blood or on a sample obtained with a small brush rubbed on the inside of a person's mouth. The test can find about 90% of non-Jewish Caucasian carriers of Northern European background, and about 97% of Ashkenazi Jewish carriers. Approximately half of the Hispanic and African-American carriers can be found. In every population some CF carriers will be missed by the test.

Is CF Carrier Testing for You?

CF carriers do not have any health problems from their one CF gene. Many people, therefore, do not become interested in whether or not they are CF carriers until they are planning to have a child. Having carrier testing before pregnancy is ideal, but if a pregnancy is already underway, testing can still be done.

Occasionally a couple with no known family history of CF decides to have CF carrier testing after discussing the test with their doctor or with a genetic counselor. If there is no one in your family who has had CF, then the chance that you are a CF carrier is about:

- 1 in 25 if you are Caucasian (White)
- 1 in 65 if you are African-American
- 1 in 150 if you are Asian

If someone in your family does have CF, then, no matter what your ethnic or racial background, your chance of being a carrier is increased. The chance is greater if the person with CF is a close relative. You should discuss your family history with your doctor or with a genetic counselor, after which you and your partner may decide to have the CF carrier test. Sometimes it is also necessary to test the relative who has CF, or other family members, to tell whether or not you are a carrier.

What if Your Test Result Shows You are a Carrier?

If your CF carrier test shows that you have one CF gene and one normal gene, then you are a CF carrier. The test has greater than 99% accuracy. This result does not affect your health in any way. Only if both you and your partner are CF carriers could you have a child with CF. It is therefore recommended that your partner also have the CF carrier test.

What if Your Test Result Does Not Show a CF Gene?

If your test result does not show a CF gene, the chance that you are a CF carrier is low. The test report takes into account your ethnic and racial background and any family history of CF. Because the test cannot find all CF carriers, the chance of being a carrier is never zero.

The results of the CF carrier tests for a couple will be used to figure out their chance of having a child with CF. If the man who was tested is not the father of the baby (non-paternity), then the real father must be tested before you can know the chance that baby will have CF.

Is There Prenatal Testing for CF?

The same CF test that is done on blood or cheek-brush samples can also be done on prenatal samples (amniotic fluid or chorionic villi). Accurate interpretation of the test depends on knowing that the man tested is in fact the father of the unborn baby. The situations where couples consider prenatal testing for CF include:

- Pregnancies where both parents are CF carriers: These pregnancies have a 1 in 4 (25%) risk for resulting in a child with CF. When both parents have CF carrier tests that show one CF gene, the prenatal test can tell with great accuracy whether the unborn baby does or does not have the genes which result in CF.
- Pregnancies with suggestive ultrasound findings: Occasionally a couple with no family history of CF finds out from an ultrasound that there is a block in the intestines in the fetus. This can be a sign of CF. The doctor may offer CF carrier tests to the parents before, or at the same time as, prenatal testing.
- Other pregnancies: Parents with other at-risk situations should talk to their healthcare professional.

How Can Genetic Counseling Help?

A genetic counselor or doctor can:

- Help you understand your chance of having a child with CF
- Explain which other family members may need to be tested
- Explain your CF carrier test results
- Explain prenatal CF testing
- Talk to you about your feelings and answer other questions

What is Cystic Fibrosis?

Cystic Fibrosis (CF) is one of the most common inherited diseases. About 1 in 2500 Caucasians (Whites) in the United States has CF. In people of other ethnic or racial backgrounds, the condition is less common.

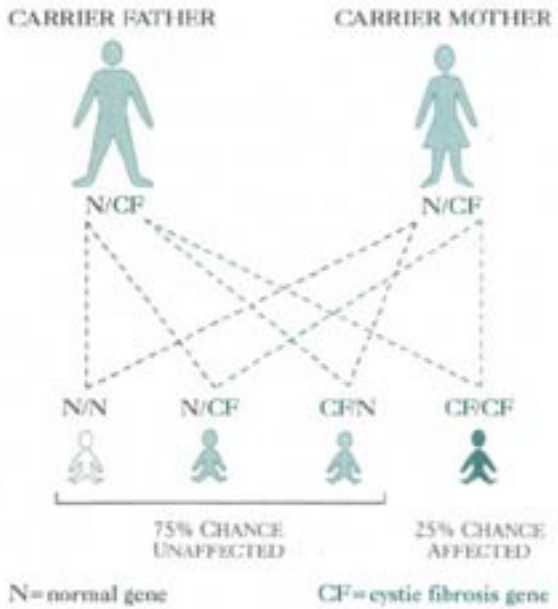
CF causes the body to produce large amounts of abnormally thick mucus. This mucus collects in the lungs, leading to congestion and pneumonia. Mucus may also collect in the intestines, resulting in diarrhea and poor growth. Treatment for these problems often requires staying in the hospital for one or two weeks at a time. Although CF is present from birth, the symptoms may not appear until later in childhood, or in some people until teenage or adult years. CF does not affect intelligence.

There is no cure for CF at this time. Scientists are, however, making progress in improving treatment and in searching for a cure. In the past, people with CF died very young, but now many are living into their late 20's or 30's. The life span of children born with CF today is expected to be even longer.

How is CF Inherited?

Cystic fibrosis results from having two CF genes. Genes are packages of information that act like blueprints for making the proteins that work in our bodies. A change in genetic information can result in an abnormal protein that doesn't work properly.

Individuals with CF have inherited one CF gene from each parent. A parent who has one CF gene and one normal gene is called a *carrier*. A carrier is not affected with the disease, but may pass to a child either the CF gene or the normal gene.



As the picture shows, in each pregnancy there is a 1 in 4 (25%) chance that the child will inherit two CF genes and will have CF. There is a 75% chance that the child will not have the disease: the child could be a carrier like the parents, or could inherit two normal genes. These chances are the same for each pregnancy, no matter how many children a couple has had. The chances are also the same for boys and girls.