Cystic Fibrosis Testing

Reasons for testing

Understanding your testing options helps you prepare for the future. You may have many reasons for testing, including if you have:

- concerns regarding inherited diseases
- an increased risk of being a carrier of an inherited disease due to ethnic background
- a family history of a specific inherited disease

Genetic disorders are caused by a change in a gene; this change is called a mutation. Every person has two copies of each gene, one inherited from each parent. A carrier is a person who has one normal copy of a gene and one abnormal copy. Having one normal gene is enough to prevent the disease. However, if both parents are carriers of the same abnormal gene, there is a chance that each parent will pass his or her abnormal gene on to their baby. If the baby inherits two copies of the abnormal gene, the baby will have the disease. Diseases that are inherited this way are called autosomal recessive diseases.

Couples may decide to have carrier testing to find out if they are carriers, and therefore are at risk of having a baby with one of these genetic diseases. If both parents are carriers of the same disease gene, prenatal diagnosis can be performed to determine whether or not the fetus is affected.

If both parents are carriers of an abnormal gene, there is a chance that each parent will pass the abnormal gene on to their child. An individual with two abnormal genes for an autosomal recessive disease will be affected with the disease. When both parents are carriers, with each pregnancy there is a:

- 25% (1 out of 4) chance that the child will inherit two normal genes, and will be neither a carrier, nor affected with the disease
- 50% (2 out of 4) chance that the child will inherit one normal and one abnormal gene, and will be a carrier of the disease (but not affected with the disease)
- 25% (1 out of 4) chance that the child will inherit two abnormal genes, one from each parent, and will be affected with the disease

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The American College of Obstetricians and Gynecologists recommends that couples planning a pregnancy, or those already pregnant, be informed about cystic fibrosis (CF) and CF carrier testing. Testing can help determine if a couple is at risk of having a child with CF.

Your risk for being a CF carrier is determined by your family history and your ethnic background. If there is no one in your family with CF, then your baseline risk for being a CF carrier is determined by your ethnicity. CF is more common among people of Caucasian and Ashkenazi Jewish descent. The disease is less common among those of Hispanic, African American, Native American or Asian descent.

If you do have a family history of CF, then your chance of being a carrier is increased above your risk based upon your ethnicity alone. The chance is even greater if the person with CF is a close relative such as a parent, sibling, or child

Ethnicity	CF Carrier Risk in People with No Family History of CF
Caucasian	1 in 25
Ashkenazi Jewish	1 in 26
Hispanic	1 in 46
African American	1 in 65
Asian	1 in 90

How the test works

Carrier testing is performed by testing a blood sample to see if a person has one abnormal copy of a specific gene. Pregnant woman are usually tested first, and if the test is positive for a carrier, then the woman's partner will be tested.

What's not detected

Carrier testing usually tests for the most common mutations that can result in an abnormal gene. Carrier testing often does not detect less common mutations.

Understanding results

A negative result significantly lowers, but does not completely eliminate, the risk of being a carrier.

For CF, if the test determines that you are a carrier, the next step is for your partner to have carrier testing performed. Both parents must be carriers for the baby to be at risk for an autosomal recessive disease. If you are determined **to be a carrier and your partner has a negative test result and no family history of CF, the chance that your baby will have CF is less than 1%.**

If carrier testing shows both parents are carriers or if testing determines that a couple is at high risk, prenatal testing (chorionic villi sampling [CVS] or amniocentesis) during pregnancy can be done to see whether or not the baby has inherited the disease. In these scenarios you will be referred to a genetic counselor for further discussion and if indicted, further testing.

We routinely order this test for first time mothers and patients who are unsure about prior testing. **If you do not want testing, please let us know *before* getting your prenatal labs drawn.**