

Cystic Fibrosis Genetic Carrier Testing



Does this test have other names?

Cystic fibrosis genetic testing

What is this test?

This test is done to see if you carry an abnormal gene that may cause cystic fibrosis (CF) in your child. A defect in a gene is called a mutation. Genes are made from DNA, and mutations can be found by doing special tests that look at your DNA.

CF is a serious disease that causes thick mucus to form in the lungs, pancreas, and other organs. CF may be treated, but the disease has no cure.

Every person has 2 copies of every gene, 1 copy from each parent. To have CF, you must have inherited a nonworking CF gene from each parent. If you have only one copy of the nonworking CF gene, you will not have the disease, but you would be a carrier of it. You could pass the disease on to your children.

Why do I need this test?

The American College of Obstetricians and Gynecologists recommends that all couples who are pregnant or thinking of getting pregnant consider carrier testing for CF. You may choose to have this test if:

- You are considering becoming a parent
- You or your partner is pregnant
- You have a family history of CF, and you want to know whether you are a carrier of the gene

Your chance of being a carrier of the CF gene is higher if you have a family history of the disease. More than 10 million Americans are carriers of this gene, and many don't know it. Your racial background has a lot to do with your risk for being a carrier. The risk is about:

- 1 in 29 for Caucasian (white) Americans
- 1 in 46 for Hispanic Americans
- 1 in 65 for African Americans
- 1 in 90 for Asian Americans

Making the decision to find out whether your child may be born with CF may be difficult. It's important to discuss this decision with a genetic counselor, your healthcare provider, your partner, family members, and possibly a counselor.

What other tests might I have along with this test?

You may have other tests to screen for diseases or conditions that could be passed on to your child. These types of tests may be included in your prenatal testing.

What do my test results mean?

Test results may vary depending on your age, gender, health history, and other things. Your test results may be different depending on the lab used. They may not mean you have a problem. Ask your healthcare provider what your test results mean for you.

Your healthcare provider or a genetic counselor will discuss your test results with you and help you understand what they mean. If your test result is positive, it says you are a carrier of the CF gene. The result is more than 99% accurate.

A negative result is not as accurate. Even if your test result says you are not a carrier, you may still have a slight chance of being a carrier for 1 of the rare types of gene mutations that do not show up on the test.

If you are a carrier but your partner tested negative for CF, there is still a slight chance that your child will be born with the disease.

It's important to know whether you carry any changes to the CF gene. You may want to let other family members know if the gene change runs in your family. If you and your partner are both carriers, this is what your tests may mean:

- Your child has a 50% chance of being a carrier of the CF gene but will not have the disease.
- Your child has a 25% chance of having CF.
- Your child has a 25% chance of not having CF or being a carrier.

How is this test done?

The test needs cells from your body so that your DNA can be studied. This may be done with a blood sample, which is drawn by putting a needle into a vein in your arm. You may also have a swab rubbed inside your mouth to get cells from inside your cheek.

Does this test pose any risks?

Having a blood test with a needle carries some risks. These include bleeding, infection, bruising, and feeling lightheaded. When the needle pricks your arm or hand, you may feel a slight sting or pain. Afterward, the site may be sore. There are no risks for the swab test.

What might affect my test results?

No medicines or conditions will affect your test results.

How do I get ready for this test?

You don't need to prepare for this test. But if you have a cheek swab, you should not eat or drink anything 30 minutes before the test. Be sure your healthcare provider knows about all medicines, herbs, vitamins, and supplements you are taking. This includes medicines that don't need a prescription and any illegal drugs you may use.

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