

The CYSTIC FIBROSIS Test

Information for
Patients

Genetic and Prenatal
Risk Assessment
for Cystic Fibrosis



LENETIX[®]
MEDICAL SCREENING LABORATORY, INC.



CYSTIC FIBROSIS Test

Commonly Asked Questions

What is Cystic Fibrosis?

Cystic Fibrosis (CF) is one of the most common inherited diseases, affecting about 1 in 3300 people in the United States. It is most common in Caucasians, but does occur in other ethnic backgrounds.

CF causes the body to produce thick mucus leading to pneumonia, diarrhea, poor growth and infertility. Intelligence is normal. While severely affected individuals die in childhood, the average life span is around thirty years; and may improve as scientists search for better treatments.

Why are Some Babies Born with CF?

If both parents are carriers of a CF gene, each of their children has a 25% chance of having the disease and a 75% chance of not having it. These chances are the same for each pregnancy, regardless of sex. Carriers are not themselves affected by the disease.

The diagram below shows the different genetic combinations that can be passed to children when both parents are carriers, each having one normal gene (N) and one CF gene (CF).

	MOM (N, N)	MOM (N, CF)
DAD (N, N)	Non Carrier N N	Carrier N CF
DAD (N, CF)	Carrier CF N	Affected CF CF



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Who are Carriers and How Can They Find Out?

If there is no one in your family with CF, your risk for being a CF carrier is shown in the following table along with the detection rates of the CF carrier test (the numbers vary slightly among different laboratories).

If You Are: ↓	Your Risk of Being a CF Carrier is:	The Detection Rate is:	Carrier Rate After Negative Test Result *
Ashkenazi Jewish	1/24	94%	1/400
Non-Hispanic Caucasian	1/25	88%	1/208
Hispanic American	1/46	72%	1/164
African American	1/65	65%	1/186
Asian American	1/94	49%	1/184

*As stated in ACOG Committee Opinion No. 325

If someone in your family has CF, then no matter what your ethnic background, your chance of being a carrier increases depending on how closely related you are to the person with CF. Your specific risk can be determined by a doctor, or by a genetic counselor.

CF carrier testing requires a small sample of blood or a swab of cheek cells. The results are available in approximately one week.





What if Your Test Results Show You are a CF Carrier?

If your test results show you have one CF gene, then you are a CF carrier.

- If one member of the couple is a CF carrier and the other is not, then the risk is less than 1 in 1000 (0.1%) that any child of theirs would have CF.
- If both parents are CF carriers, each pregnancy has a risk of 1 in 4 (25%) that the child will be affected.
- If only one member of the couple has been tested and is found to be a carrier, the partner should be tested as soon as possible.

The information from the carrier test may help in your family planning, as well as alert family members to the possible need for CF testing. You may wish to consult with your doctor or a genetic counselor for further discussion of what the results mean for you and your family.

What if Your Test Results Do Not Show a CF Gene (screen negative)?

CF Testing cannot find all carriers. The test takes into account your ethnic background and any family history of CF. If your test results do not show a CF gene, the chance that you are a CF carrier is low, but never zero. Not all changes (mutations) in the CF gene are detected by CF carrier testing.

Is There Prenatal Testing for CF?

Yes. If both parents are shown to be carriers, then prenatal testing by chorionic villi sampling or amniocentesis can be performed to determine whether or not the fetus is affected with CF, a carrier of CF, or a non carrier. The accuracy of the prenatal test is greater when both parents have been tested for CF. The accuracy also depends on knowing that the man tested is in fact the biological father.

Do I Have To Have a CF Test?

It is your decision whether or not to undergo CF testing.

When Should I Consider Prenatal Testing for CF?

Prenatal testing for CF is appropriate when:

- Both parents of the pregnancy are CF carriers
- Ultrasound (sonogram) of the fetus shows signs of CF, such as thickened intestines. In this case, blood samples or cheek swabs from both parents should be sent for testing at the same time as the CVS or amniocentesis specimen.
- There is any other at-risk situation. Talk to your doctor or to a genetic counselor.

Will Insurance Cover the Cost of the CF Carrier Test?

Health plans and insurance coverage may vary depending on policy, plan or state regulations. It is best to ask your plan representative regarding test coverage prior to having the carrier test. Tests performed without coverage are billed directly to the patient and are the patient's responsibility.

For Additional Assistance

This brochure contains general information regarding carrier testing for CF. However, you may wish to obtain professional genetic counseling prior to having the test.

The individual capabilities and potential of children with CF are considerations which you may wish to discuss with your counselor or with other healthcare providers. Further information and support are available through groups such as your local Cystic Fibrosis Association.

CYSTIC FIBROSIS INFORMED CONSENT

I have read and understand the information in this pamphlet regarding screening for the Cystic Fibrosis test.

- Yes, I want to have Cystic Fibrosis testing.
 No, I do not want to have Cystic Fibrosis testing.

Patient Name: _____

Patient Signature: _____

Date: _____ **IMPORTANT: Retain Copy in Patient File**



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The information included in this pamphlet is not intended as a substitute for personal medical advice. Specific situations always require a personal consultation with your healthcare provider.

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