Cystic fibrosis carrier screening

Why is carrier screening for cystic fibrosis important?

Carrier screening tests help identify individuals who may have an increased risk of having a baby with certain genetic conditions. Even if you are healthy, have no family history of the condition, or even already have healthy children, you may be a carrier of a genetic condition. One of these conditions is cystic fibrosis.

This brochure will provide you with information to help you learn more about cystic fibrosis carrier testing.

The ACOG Committee on Genetics recommends cystic fibrosis carrier screening be offered to all patients who are planning a pregnancy or seeking prenatal care because it is becoming increasingly difficult to assign a single ethnicity to individuals.¹

Test limitations

No test is perfect. While results of the HerediT® Cystic Fibrosis Carrier Screen test are highly accurate, a negative result significantly reduces but does not eliminate the chance of being a carrier. The results of this testing, including the benefits and limitations, should be discussed with your healthcare provider.

Sequenom Laboratories, a wholly-owned subsidiary of Sequenom, Inc., is a CAP-accredited and CLIA-certified molecular diagnostics laboratory dedicated to improving patient outcomes by offering revolutionary laboratory-developed tests for a variety of prenatal and eye conditions. Sequenom Laboratories pioneered NIPT for fetal aneuploidies with the launch of its MaterniT21™ PLUS test, and offers a full menu of prenatal tests.

The HerediT Cystic Fibrosis carrier screen test is a laboratory-developed test that was developed, validated and is performed exclusively by Sequenom Laboratories.

References


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What is cystic fibrosis?

Cystic fibrosis (CF) is one of the most common genetic conditions in the United States. It is caused by changes in the CFTR gene. Changes in this gene cause the body to produce thick sticky mucus in the lungs, pancreas and other organs that can affect breathing and digestion. CF does not affect everyone the same way, therefore, some people may be more severely affected than others. Symptoms can range from moderate to severe and can even impact fertility. The average lifespan of someone affected with CF is 37 years. It is estimated that more than 10 million Americans are carriers of CF. While the risk of being a CF carrier is dependent upon one’s ethnicity and family history, individuals of all racial and ethnic groups may be carriers of CF.

Patients who may benefit from a CF carrier screen test:

- Couples considering having a child or those already pregnant
- Patients with a history of CF or male infertility
- Patients who have a reproductive partner who is a CF carrier
- Ultrasound findings that indicate an increased risk for CF

The HerediT® Cystic Fibrosis carrier screen test is a simple blood or buccal (cheek) swab test and results are typically available to your health care provider within one week.

How is cystic fibrosis inherited?

CF is inherited in a recessive pattern. This means that, in order to be affected, a person must inherit two disease-causing mutations—one from each parent. To be at-risk to have an affected child, both parents must be carriers of one of the CF disease-causing mutations. Carriers have only one mutation and usually have no symptoms of CF.

If both parents are carriers, there is a one-in-four (25%) chance with each pregnancy that a child will have CF, and a one-in-two (50%) chance that a child will be a CF carrier. It is estimated that one in 30 Americans are carriers of CF.

What does a positive carrier screening test result mean?

A positive CF carrier screening test result means you have one copy of a mutation that is known to cause CF. It does not mean you have CF. If you are found to be a CF carrier, then your partner should be tested. If you are both found to be CF carriers, your doctor, genetic counselor or other health care provider will discuss reproductive and prenatal testing options with you.

What does a negative carrier screening test result mean?

A negative CF carrier screening test significantly reduces your risk to be a CF carrier, but it does not reduce your risk to zero. Because this test does not look for all CF mutations, and because not all CF mutations may even be known at this time, a negative result can’t completely eliminate the chance that you could be a carrier.

For more information, ask your healthcare provider or visit sequenom.com