

# *Cystic Fibrosis Carrier Testing*

## **Introduction**

Cystic fibrosis carrier testing before or during pregnancy can help determine your risk of having a child with cystic fibrosis. This information explains cystic fibrosis and cystic fibrosis carrier testing.

## **What is cystic fibrosis?**

Some background information about cystic fibrosis (CF) is useful as you decide whether to pursue CF carrier testing.

CF is a lifelong, genetic (inherited) condition that affects the production of mucus, sweat, saliva, digestive juices and sperm. Normally, these secretions are thin and slippery. However, for people who have CF, the secretions can become thick and sticky and may build up, especially in the lungs, pancreas and intestines. This can lead to respiratory (breathing) and digestive problems.

CF is not contagious and does not affect a person's appearance or intelligence. It usually is diagnosed in early childhood, although milder forms of the disease may not be detected until later in life. It affects both males and females.

The average life span of a person with CF is around 30 years, although some people live longer. Research is ongoing to develop improved treatments and help people with CF live longer, more comfortable lives.

## **What are the symptoms of cystic fibrosis?**

The symptoms of CF vary. Some people have only breathing problems, some have only digestive problems and some have both. Symptoms may get worse with age.

Typically, CF causes respiratory problems, such as chronic cough and wheezing, frequent chest and sinus infections and chronic bronchitis. Respiratory failure (stopping breathing) is the most dangerous result. Typical digestive difficulties include chronic diarrhea, intestinal blockage and malnutrition.

Many people who have CF lose too much salt when they sweat, causing an imbalance of minerals in the blood. CF also may lead to diabetes, liver damage (cirrhosis), growth delays, rounding (clubbing) of the fingertips and toes and infertility (especially in men).

A rare form of CF causes only mild respiratory or digestive symptoms and may not be diagnosed until adulthood.

## **How is cystic fibrosis treated?**

Currently, CF cannot be cured. Treatment focuses on managing symptoms and preventing complications of the disease. A person who has CF should have regular appointments with a health care provider to monitor symptoms and evaluate the response to medication and other treatment.

Most people who have CF need daily respiratory therapy, such as special exercises to loosen and promote drainage of thick, sticky mucus from the lungs. Medication may be prescribed to help thin mucus and clear the airways. Antibiotics can help fight lung infections. Over time, lung infections tend to become more frequent and serious, and may require treatment in a hospital.

Digestive problems typically are treated with medication (digestive enzyme supplements) and vitamins.

## **What causes cystic fibrosis?**

CF is an inherited disease, passed from parent to child through the genes. Genes contain the basic instructions that tell each cell how to function. Mistakes or alterations (mutations) in genes can keep cells from working correctly and lead to genetic diseases, such as CF. CF does not result from anything a person or his or her parents did or did not do before, during or after pregnancy.

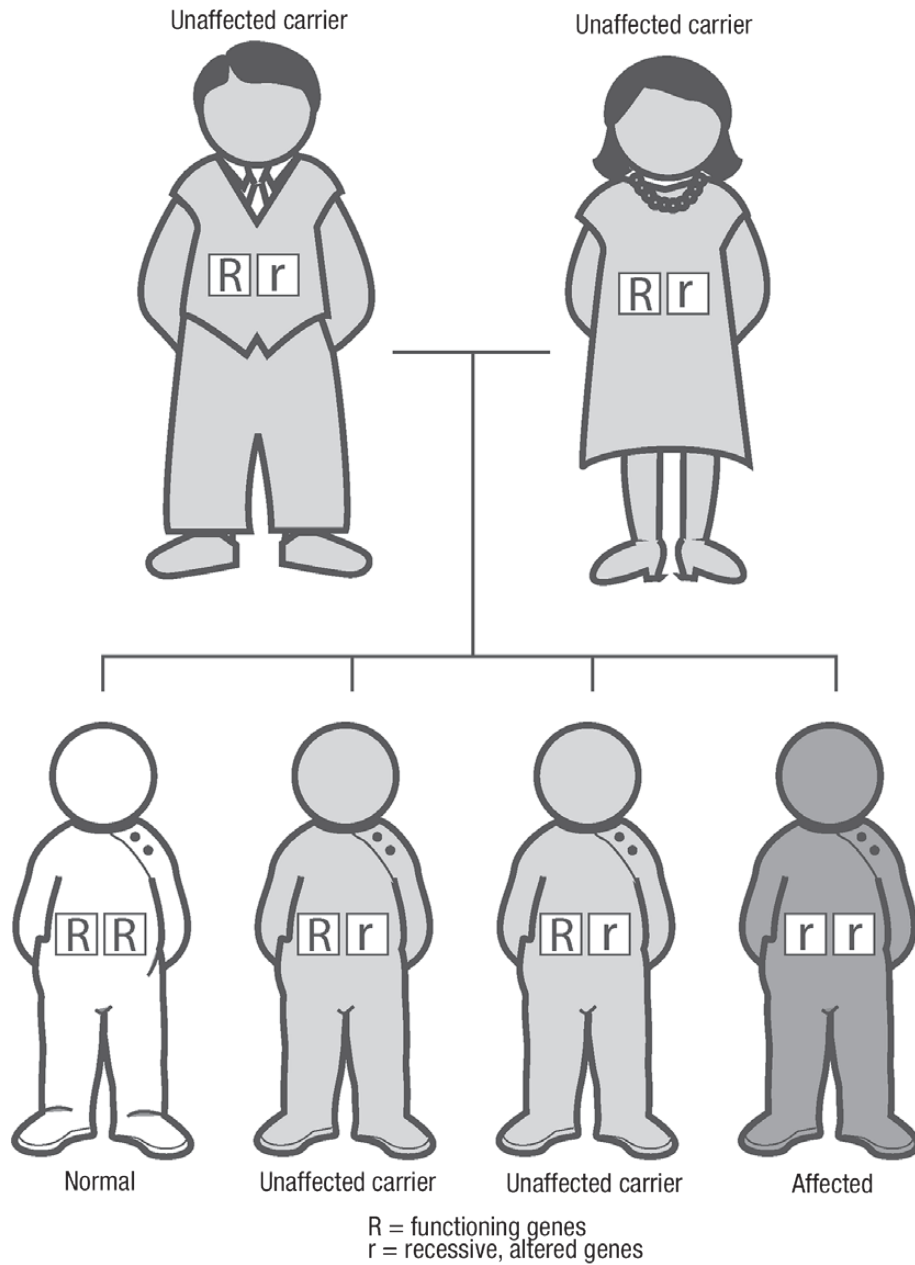
In CF, a genetic mutation gives faulty instructions for moving salt and fluid in and out of the body's cells. This leads to abnormal secretions, particularly in the airways and digestive tissues, and causes the symptoms of CF.

## **How is cystic fibrosis inherited?**

CF is passed from parent to child through the genes. Genes come in pairs. A child receives one copy of each gene from the mother, and one copy from the father. To develop CF, both copies of the gene for CF must be altered. A person who has only a single altered gene for CF will not have CF, but is an unaffected carrier and could pass the genetic alteration to his or her children.

As figure 1 shows, when both parents carry a single altered CF gene, each child has:

- A 25 percent chance of having CF.
- A 25 percent chance of not having CF or being a carrier of the disease.
- A 50 percent chance of carrying a single altered CF gene.



**Figure 1.** Chances of inheriting cystic fibrosis when both parents are carriers

## Who gets cystic fibrosis?

Certain factors may increase the risk of having a child with CF. The greatest risk factor is a family history of the disease. If either partner comes from a family with a history of CF, the chance increases of being a carrier and having a child with the disease.

Ethnicity plays a role as well. While people of any ethnic background can carry CF, the risk is higher among certain groups (see “Ethnicity and Cystic Fibrosis” below).

*\*Reprinted with permission from the The American College of Obstetrics and Gynecologists.*

<i>Ethnicity and Cystic Fibrosis*</i>		
<b>Ethnicity</b>	<b>Chance of being a CF carrier</b>	<b>Chance of having a child with CF</b>
Northern European/Caucasian	1 in 25	1 in 2,500
Ashkenazi Jewish	1 in 25	1 in 2,500
French Canadian	1 in 25	1 in 2,500
Hispanic/Latin American	1 in 46	1 in 8,464
African American	1 in 65	1 in 16,900
Asian American	1 in 90	1 in 32,400

## What is cystic fibrosis carrier testing?

CF carrier testing can provide information about your risk of being a carrier of CF and can help determine your chance of having a child with the disease. If you choose to have the test, a sample of your blood will be tested for specific alterations in the gene for CF. The test looks for the most common genetic alterations associated with CF, not for every possible alteration (see “What are the limitations of carrier and prenatal tests?”). This test is available from Mayo Clinic Laboratories as Mayo ID: CFP / Cystic Fibrosis Mutation Analysis, 106-Mutation Panel, and can be ordered by your physician.

## Should I have cystic fibrosis carrier testing?

Your decision to pursue CF carrier testing involves many factors. Your health care provider can give you information about the test. He or she also may refer you to a genetic counselor — a health care professional trained to help you understand inherited conditions. A genetic counselor will review your family history, explain genetic testing (including the limitations and benefits of testing) and discuss other factors to consider when making your decision. Ask enough questions to be certain about your decision.

*When considering CF carrier testing, think about:*

- **Your individual risk.** Do you or your partner have a family history of CF? Is either of you a member of a high-risk ethnic group?
- **Family planning.** Would the test results influence your plans to have children?
- **Pregnancy management.** Would you consider having further tests on the developing fetus if you and your partner were found to carry a genetic alteration for CF?
- **Changes to your home and lifestyle.** Would you use the information to help prepare for the possibility of raising a child with special medical needs?
- **Your religious or spiritual beliefs.** Would your religious or spiritual beliefs impact your choices about prenatal testing, or continuing or terminating a pregnancy?
- **Financial considerations.** Is the test covered by your insurance policy? Some policies cover genetic testing, others do not. Check with your medical insurance carrier to determine whether your policy will pay for testing and related appointments.

## What are the limitations of carrier and prenatal tests?

Be sure to discuss the limitations and benefits of CF carrier testing with your health care provider or genetic counselor. CF carrier testing is not 100 percent predictive. CF is linked to hundreds of alterations in the CF gene, most of them rare. Current testing does not look for every possible alteration, only the most common ones. In addition, not all CF alterations are known. Even if your test is negative, you may carry an untested, undetected or unknown genetic alteration that increases your risk of having a child who has CF.

If prenatal tests show your developing baby has CF, you may not know how the disease will affect him or her. Genetic testing usually cannot predict the severity of CF.

## What do my test results mean?

This section outlines the general meaning of various carrier test results. **Discuss the results of your test with your health care provider or a genetic counselor.** (Ask your health care provider when and how you will receive the test results.)

- **Your test is negative.** A negative result means that none of the common genetic alterations associated with CF were detected, and your chance of having a child who has CF is significantly reduced. Even if your partner receives a positive test result, both of you must carry an altered CF gene to pass the condition to a child.

However, remember that no test is an absolute guarantee (see “What are the limitations of carrier and prenatal tests?”). Following a negative test result, your health care provider or genetic counselor may give you information about any remaining risk of carrying CF.

- **Your test is positive.** A positive result means that you carry an altered gene for CF. Being a carrier does not put you at risk of any health complications associated with CF. However, as a carrier, you are at risk of having a child who has CF if your partner also carries an altered CF gene (see “Both your and your partner’s tests are positive”).

If your test is positive, your partner should consider carrier testing. If your partner’s test is negative, your chance of having a child with CF is very small. (Again, no test is an absolute guarantee.) If you have a new partner for a future pregnancy, he or she should consider carrier testing as well.

Both your and your partner’s tests are positive. If both of you receive positive test results, you have a 25 percent chance of having a child who has CF, a 50 percent chance of having a child who carries a genetic alteration for CF and a 25 percent chance of having a child with no genetic alterations for CF. Your health care provider or genetic counselor can give you information and help you evaluate your options. You may decide to have children; you may explore a pregnancy using donor sperm or eggs; you may decide not to have children; or you may decide to pursue adoption.

Preimplantation genetic diagnosis (PGD) is another option available at some hospitals and medical centers. PGD involves obtaining embryos by fertilizing eggs with sperm in a laboratory (in vitro fertilization). Embryos are tested for genetic disorders before they are transferred to the uterus (womb). Your health care provider or genetic counselor can discuss the benefits and risks of PGD and help you decide whether it is an option for you.

If you and your partner are both carriers for CF and you choose to conceive or are already pregnant, your health care provider or genetic counselor can help you decide whether to have prenatal testing on your developing baby. CF cannot be treated before birth, so the purpose of these tests is to help you plan for the future.

## Contacting your health care provider

If you have questions about CF carrier testing or this information, contact your health care provider or genetic counselor.

*This material is for your education and information only. This content does not replace medical advice, diagnosis or treatment. New medical research may change this information. If you have questions about a medical condition, always talk with your health care provider.*

MAYO CLINIC LABORATORIES | 3050 Superior Drive NW | Rochester, MN 55901 | [mayocliniclabs.com](http://mayocliniclabs.com) | 800-533-1710

©2018 Mayo Foundation for Medical Education and Research. All rights reserved. MAYO, MAYO CLINIC, Mayo Clinic Laboratories and the triple-shield Mayo logo are trademarks and service marks of MFMER.