Frequently Asked Questions About Genetic Carrier Testing for Cystic Fibrosis (CF)

Medical technology now offers information about the future health of individuals. Using genetic information, tests can be offered to find out if a person might have a child who may have certain diseases or health care needs.

This fact sheet addresses questions about genetic carrier testing for cystic fibrosis (CF). Genetic testing looks at the genes that a person has to learn if that person carries an altered gene for CF. An altered gene also may be called a CF mutation. For someone to have this disease, two copies of an altered CF gene must be present-one from each parent. A "carrier" has only one copy of an altered CF gene and therefore does not have CF.

The Cystic Fibrosis Foundation is focused on finding a cure for this disease, improving the quality of life for the 30,000 people in the US with CF and sharing information about living with this condition. The CF Foundation does not make specific recommendations about who should have these tests. If you would like more information, you should speak with your doctor or a genetic counselor about the test.

What is genetic carrier testing for CF?

Genetic carrier testing can be used to tell if a person carries one of the altered genes that causes CF. The test looks at a person's DNA (genetic material), which is taken from cells in a blood sample or from cells that are gently scraped from inside the mouth. There are more than 1,000 known alterations of the gene that causes CF. Current tests look for the most common CF gene alterations. A negative test means the person does not carry most of the alterations, but the person still may carry a rare CF gene.

The altered genes that are looked for by the test vary according to a person's race or ethnic group, or if CF occurred already in the family. About 95 percent of Americans with CF are white or Caucasian. One person in every 29 people who are Caucasian are carriers of an altered CF gene. In other race or ethnic groups, one in 46 Hispanic Americans, one in 65 African Americans and one in 90 Asian Americans carry an altered CF gene. If you have a relative with CF, or who is known to carry the altered CF gene, your chance of carrying the gene is greater because of your family's history. **If you are pregnant or planning to have a child, you should discuss this test and the results with a health professional who is knowledgeable about genetic testing, such as a genetic counselor.**

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Cystic Fibrosis Foundation

Genetic Carrier Testing for CF

What is CF?
Cystic fibrosis is a genetic disease that causes the body to make thick, sticky mucus. This is because of the faulty movement of sodium and chloride (the ingredients of salt) in the cells of certain organs, such as the lungs and pancreas. Symptoms of CF include: salty-tasting skin; coughing, wheezing and recurrent lung infections; good appetite but poor weight gain; and bulky bowel movements. In the lungs, this mucus leads to chronic infections and increasing loss of lung function. In the pancreas, the thick mucus keeps enzymes from reaching the gut to digest food. The disease also affects the ability of most men with CF to have children.

There are about 30,000 children and adults with CF in the United States. Because of improved medical treatments, the median life expectancy for a person with CF is in the early 30s. And, children diagnosed with CF today may live into their 40s or older. Now, more than one third of people with CF are adults of all ages. The outlook for people with CF is improving; however, there is no cure. To learn more about CF and how it is treated, contact the CF Foundation at **(800) FIGHT CF**, or visit the CF Foundation's Web site at **www.cff.org**.

Should I get a test to see if I am a carrier of the CF gene?
Your decision to get a genetic test to learn if you carry an altered CF gene is a difficult and personal choice. You may want to talk to your medical or religious advisors to help you decide. The American College of Obstetricians and Gynecologists (ACOG) suggests that all couples who are thinking about having a baby -- or those who are already pregnant -- should get genetic carrier testing for CF. Although Caucasians have a higher risk of carrying the altered CF gene, ACOG favors testing for anyone who requests it. To learn more about genetic carrier testing in general, visit ACOG's Web site: **www.acog.org**.

How does someone inherit the altered CF gene?
People inherit genes from their parents. To inherit CF, a child must get one copy of the altered CF gene from each parent. In other words, the child must have two copies of the altered gene to have CF. A person who has only one altered gene does not have CF, but is considered a "carrier" of the CF gene. Both males and females may inherit the disease.

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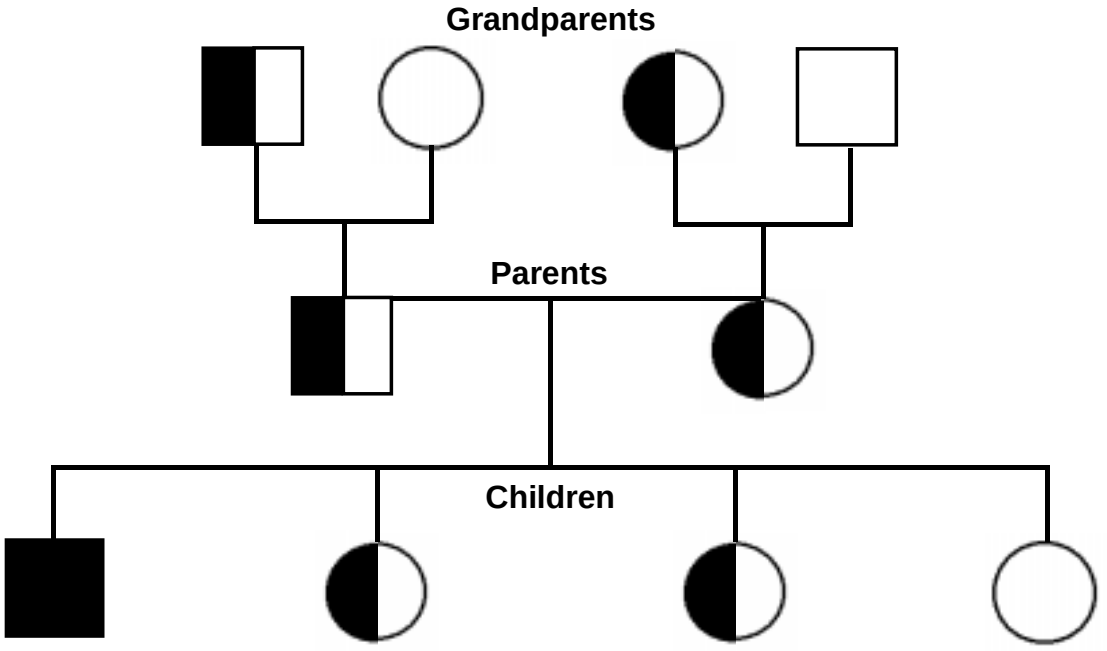
Inheriting Cystic Fibrosis—A Gene Carrier Chart



What do the results of a genetic carrier test for CF mean?
A positive genetic carrier test for CF means that a person has an altered copy of the CF gene; this result is more than 99 percent accurate. A "negative" carrier test, which says that someone does not carry the CF gene, is not as accurate. With more than 1,000 different alterations or mutations of the CF gene, there are some rare ones that the test does not identify. If your test is negative for an altered CF gene, there is still a small chance you could be a carrier of one of these rare mutations. This chance depends on your race or ethnic group and the type of genetic carrier test you receive.

If I have one copy of the altered CF gene, can my children have CF?
Yes, if you have one copy of the altered CF gene, your child may still have CF. However, your child will only have CF if your partner also is a carrier of an altered CF gene. If you are both carriers, each baby you have together has a 25 percent chance of having CF. If one partner is a carrier and the other partner tests negative for the altered gene (or if there is no information on whether or not he or she carries the altered gene), there is still a slight chance the child could have CF. Your doctor or a genetic counselor can tell you about your chances of having a child with CF.

How can I carry a CF gene when no one in my family has ever had the disease?
Since one copy of an altered CF gene does not cause symptoms, this copy can be passed down to family members without any impact on their health. Unless they have a child with CF, most people who carry an altered CF gene do not know they carry it. (See diagram.) Once

parents have a child with CF, all of their children should be tested for CF. Other relatives, such as brothers, sisters or cousins, have a chance of being CF carriers and might want to have carrier testing.

Do certain types of the altered CF genes make the disease more mild or severe?
Yes, the type of altered CF gene can affect the type of symptoms of CF. But, genetic testing cannot tell how severe a person's CF will be in advance. The most common altered gene, delta F508, causes the most common CF symptoms. But, this altered gene can vary in how severely it affects someone with CF. Some less common mutations of the CF gene may cause milder symptoms.

Can genetic testing diagnose CF?
Genetic tests can diagnose CF, but these tests are often used to find out if a person carries an altered gene. The standard test to diagnose CF is called the sweat test. It measures the salt in a person's sweat. People with CF have saltier sweat than most people. If someone is diagnosed with CF through a sweat test, a genetic test can show what the altered CF genes are. To learn more about the sweat test, visit the CF Foundation's Web site at: http://www.cff.org and look under the "Living with CF" section, or call the CF Foundation at **(800) FIGHT CF**, and ask for the "Sweat Testing Fact Sheet."

Can a baby be tested for CF before birth?
Yes, a baby can be tested for CF before birth. If the mother and father are both carriers of altered CF genes, or if the mother is a carrier, genetic testing for CF can be done before birth. To learn more, ask your doctor, obstetrician, midwife, or genetic counselor.

Is there hope for a healthy future for people with CF?
When scientists found the CF gene in 1989, they gained a great tool for finding new CF treatments and, someday, a cure. There is exciting research to change the gene that causes CF, and other research to fix the symptoms of CF. Today, there are new treatments that help many people with CF to live full, active lives, and more treatments are coming in the future.

If you would like to learn more about CF and its treatments, visit **www.cff.org** or call the CF Foundation at **(800) FIGHT CF**. The CF Foundation also can help you to identify a local CF care center.

Other Resources
The American College of Obstetricians and Gynecologists: (800) 762-2264 or **www.acog.org**
National Society of Genetic Counselors: (610) 872-7608 or **www.nsgc.org.**

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