Cystic Fibrosis

A guide to carrier testing

At University of Washington Medical Center, we partner with our patients and families in making decisions about their health care.

This handout gives information to help you decide if you want to have a test to see if you are a carrier of cystic fibrosis.

Having this test is up to you. Some people do not find this type of screening test to be helpful. You may refuse testing at any time.

Talk with your health care provider to learn more.

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited, lifelong condition that causes problems with breathing and often with digestion. CF does not affect how smart someone is or how they look. There is no cure for CF, but there are many therapies and treatments for the symptoms. The disease often gets worse over time and becomes harder to treat, but many people with CF live into their 40s and older.

What causes cystic fibrosis?

When a person inherits an abnormal CF *gene* (see sidebar on the next page) from **both** parents, their body makes thick, sticky mucus. This mucus clogs the lungs, leading to life-threatening lung infections. In most people with CF, the mucus also blocks the pancreas, which keeps the body from digesting food properly.

A person who has one normal CF gene and one abnormal CF gene is known as a *carrier* of CF. Carriers are healthy and have no symptoms of CF. If two carriers of CF have a child, there is a 25% (1 out of 4) chance that the child will have CF.

What is the chance of being a CF carrier?

Cystic fibrosis occurs most often in people whose ancestors came from northern European countries, such as Ireland, England, or Holland. But, anyone can be a carrier of CF, as shown in this chart:

Ethnic Group	Chance of Being a Carrier*	
Eastern European (Ashkenazi) Jewish	4 %	(1 out of 24)
Non-Hispanic Caucasian	4 %	(1 out of 25)
Hispanic American	2 %	(1 out of 46)
African American	1.5%	(1 out of 65)
Asian American	1 %	(1 out of 94)

** From ACOG Committee Opinion 325, December 2005*

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Genes are the hereditary instructions that tell your body how to grow and develop. We have 2 sets of genes, one inherited from our mother and the other from our father. Each gene is part of a pair.

Most CF carriers do **not** have family members with cystic fibrosis. If someone in your family does have CF, your chance of being a carrier is probably higher than other people in your ethnic group. Be sure to tell your health care provider about your family's health history.

How can I find out if I am a CF carrier?

To have your CF genes tested, you will need to give a small blood sample. The lab checks for the most common changes, known as *mutations*, in the CF gene. Over 1,600 mutations in the CF gene have been discovered, but the standard test looks for the 23 mutations that are most common.

CF carrier testing is most reliable for Caucasians, because that is the group of people who usually have CF. Carrier testing is less reliable in people of other ethnic groups, as shown in this chart:

Ethnic Group	Accuracy of Carrier Test*
Eastern European (Ashkenazi) Jewish	94%
Non-Hispanic Caucasian	88%
Hispanic American	72%
African American	65%
Asian American	49%

** From ACOG Committee Opinion 325, December 2005*

What does a "negative" test result mean?

A negative test result means you do not have any of the CF gene mutations you were tested for. It does **not** mean that you're not a carrier, although chances are lower that you are. You could have one of the rare gene mutations that was not searched for.

What does a "positive" test result mean?

A positive test result means that you are a carrier of cystic fibrosis. There are no "false positives" with this test. If your CF test is positive, your partner can then be tested to see if you are both CF carriers.

What if my partner's test is negative?

If your partner's test is negative, it means his chance of being a carrier of CF is lower, but not zero. There is still a very small risk that your child will have CF. No other testing can be done during your pregnancy to diagnose CF in the baby, but your baby will be screened for CF at birth as a part of newborn screening.

Questions?

Your questions are important. If you have questions or concerns about cystic fibrosis carrier testing, talk with your health care provider before signing any consent forms.

UWMC clinic staff is also available to help at any time.

Prenatal Diagnosis Clinic:
206-598-8130

Maternal and Infant Care Clinic:
206-598-4070

What if my partner's test is positive?

If your partner's test is positive, there is a 25% (1 out of 4) chance your baby will have CF and a 75% (3 out of 4) chance your baby will not have CF.

Newborn screening does not pick up all cases of CF. If you and your partner are both carriers of CF, you should not rely on newborn screening results to diagnose CF.

Amniocentesis and *chorionic villus sampling* (CVS) are 2 procedures that can be done while you are pregnant to test your baby for CF. Or, a small blood sample can be taken after birth for genetic testing to diagnose CF.

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07/2003 Rev. 02/2009
Reprints: Health Online