

SO I HAVE A CYSTIC FIBROSIS GENE, BUT MY PARTNER'S TEST WAS NEGATIVE

You have had a cystic fibrosis (CF) carrier test that showed you are a carrier. But your partner's test was "negative." Now what?

How do you know I am a carrier for CF?

The blood test looked for the most common changes in the pair of CF genes. Since you have one of these common changes in one of these genes, you are a carrier.

What is a carrier?

A carrier is someone who has one changed gene (mutation) for the disease CF. Genes do not change during one's lifetime, so a carrier will always be a carrier, but will never get sick with CF. Carriers do not need any special medical care. People with CF always have two changed genes.

What is CF?

CF is a disease that causes breathing and digestion problems. Symptoms usually begin in the first year of life and get worse over time. Some children are very sick; others are not. Problems may include coughing, repeated pneumonia, lung damage, diarrhea and poor growth. People with CF are not mentally retarded. Their appearance is not affected. Life is usually shortened, but most children with CF live 20 to 35 years. There is no cure. Treatment is usually medicine and physical therapy.

My partner's test is negative. Can he still be a carrier?

Yes. Although the chance your partner is still a carrier is small, it is never zero. More than 900 different changes can happen in the CF gene. The test only looks for about 25 of these. Some people have a change the test does not look for. Since no change was found, there is only a small chance your partner is still a carrier. The exact chance depends on your partner's race. Your health care provider or a genetic counselor can figure out this chance for you.

What is the chance I will have a baby with CF?

Your chance to have an affected baby is small. Two things have to happen for you to have a baby with CF. First, both you and your partner must be carriers. Then, the baby must inherit the changed CF gene from both you and your partner. When both parents are carriers, the chance the baby will be affected is 1 in 4 (25%). Your chance of having a baby with CF will certainly be smaller than 1 in 4, and is probably less than 1%. Your health care provider or a genetic counselor can figure out this chance for you.

Can we test the baby?

During pregnancy:

Yes, but the results will not be very accurate. If your partner has a changed gene that the test does not look for, the changed gene will not be found in the baby either. You should talk to a genetic counselor to understand more about what the test on the baby could tell you, as well as the chance the test could cause a problem in your pregnancy.

At birth:

Yes, but again, the results will not be very accurate.

A few months after birth:

Yes. The baby can have a different test for CF that will be more accurate. This test is usually done when a baby has some symptoms of CF. You should talk to your health care provider about if and when your baby should have this test.

Should I tell other people in my family?

Since you are a carrier for CF, other people in your family may be too. You could suggest they talk to their health care provider or a genetic counselor to see if they want to be tested.

How can I get more information? How can I decide what to do?

Talk to your health care provider or see a genetics specialist, a genetic counselor. A genetic counselor is specially trained to help you decide what is best for you. A genetic counselor will answer your questions about the blood test results, the testing you could have in the pregnancy and answer any other questions you have about CF.

This fact sheet was written by the PacNoRGG (Pacific Northwest Regional Genetics Group) Education and Prenatal Genetics committees and is consistent with the 2001 Clinical and Laboratory Guidelines, *Preconception and Prenatal Carrier Screening for Cystic Fibrosis*, published by the American College of Obstetricians and Gynecologists and the American College of Medical Genetics. More detailed patient brochures, *Cystic Fibrosis Carrier Testing: The Decision is Yours*, and *Cystic Fibrosis Testing: What Happens If Both My Partner and I Are Carriers?* can be purchased from ACOG, <http://www.acog.org> 202/863-2518.

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This brochure is available on the PacNoRGG web site
<http://mchneighborhood.ichp.edu/pacnorgg>

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