

MY PARTNER AND I ARE BOTH CARRIERS FOR CYSTIC FIBROSIS

You and your partner have each had cystic fibrosis (CF) carrier testing and it showed both of you are carriers. What does this mean and what do you do next?

What is a carrier?

A carrier is someone who has one changed gene (mutation) for the disease CF. Genes don't 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.

How do you know I am a carrier for CF?

The blood test looks for the most common changes in the pair of CF genes. If you have a change in one of those genes, you are a carrier. Since you and your partner each have one of the common changes in one of the CF genes, you are both carriers.

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

When both parents are carriers for CF, the chance the baby will have CF is 1 in 4 (25%). This is the chance for every pregnancy with the same partner.

Can we test the baby during the pregnancy?

Yes. Amniocentesis and CVS are tests done during pregnancy that can check the baby's CF genes. Amniocentesis takes some of the fluid from around the baby. CVS takes a small piece of the placenta. Either of these tests will tell us if the baby has 0, 1, or 2 changed genes. A baby with 2 changed genes will have CF. A baby with 1 changed gene is a carrier and will not be sick. A baby with 0 changed genes does not have CF and is not a carrier.

What happens to a baby with CF?

Most babies with CF have problems with breathing and digestion. Symptoms usually begin in the first year of life and get worse over time. Breathing problems include coughing, bronchitis, and repeated pneumonia. Children can look underfed because of poor digestion. Children with CF do not have mental retardation and their appearance is not affected.

Some children are very sick and die in childhood. Some are not very sick and live into adulthood. We cannot predict how sick any baby will be.

There is no cure. Treatment is medicines to prevent lung infections and improve digestion, and physical therapy.

Can you predict ahead of time how sick a baby with CF will be?

No. A baby with 2 changed genes will have CF. However, some people with CF are very sick, while others are only mildly ill.

Should I have the baby tested during pregnancy?

It's up to you. This is your decision. If you want to know, have the test.

If the baby does not have CF, it will be reassuring. If the baby does have CF, there is no cure. There is no treatment before birth. You can choose to have an abortion or continue the pregnancy.

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 all your questions about your blood test results, the testing you could have during the pregnancy and 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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