ADAMS PATTERSON
GYNECOLOGY & OBSTETRICS
WOMEN'S CARE CENTER OF MEMPHIS, MPLLC

What You Should Know About Carrier Testing for Cystic Fibrosis

What is cystic fibrosis (CF)? CF is a genetic disorder usually diagnosed in the first few years of life. CF causes thick sticky mucous in the lungs which causes persistent coughing, wheezing and frequent lung infections. It also causes difficulty with digestion due to a lack of normal digestive enzymes. These problems require chronic treatments. Symptoms may be mild or severe. Men with CF are often infertile. CF does not affect intelligence or appearance. 1 out of every 3,300 Caucasian babies will be born with CF. The risk is less for other ethnic groups: 1 in 8,400 Hispanic Americans, 1 in 14,000 African Americans, and 1 in 32,000 Asian Americans. The disease course of CF is variable. Some individuals have very few medical problems and with modern treatments, survive well into adulthood.

How is CF treated? Although CF is not curable, treatments help reduce symptoms and prolong lifespan. Respiratory therapy and antibiotics help with lung problems and other medications help with digestion. These treatments usually need to be continued lifelong. In spite of therapy, people with CF usually have a shortened lifespan. Some die in childhood but many live into their 30's and 40's. Much research is being done to foster better treatments and possible cures.

What is CF carrier screening? Carrier screening tells you if you may carry the CF gene. It helps you determine what your chance of having a child with CF may be. To have a child with CF **both** you and your partner would need to carry the CF gene. Your child would need to receive the CF gene from each parent in order to be affected. If he received the gene from only one parent, he would be considered a carrier but he would not have CF.

If the test shows that you do not have the gene, could you still have a child with CF? Yes. This is because not all types of CF genes will be detected by the test. The test will detect about 80% of CF genes in Caucasians and about 97% in the Ashkenazi Jewish population. 1 in 29 persons in these groups will be found to carry the gene. The following table describes the chances of having an affected child under different scenarios:

Ethnic Group Of Both Partners	No Test	One Partner Tests Negative, One Partner Untested	One Partner Tests Positive, One Partner Tests Negative	One Partner Tests Positive, One Partner Untested	Both Partners Test Negative	Both Partners Test Positive
Ashkenazi Jewish	1 in 3,300	1 in 107,880	1 in 3,720	1 in 116	1 in 3,459,600	1 in 4
European Caucasian	1 in 3,300	1 in 16,240	1 in 560	1 in 116	1 in 78,400	1 in 4
Hispanic American	1 in 8,8464	1 in 19,320	1/420	1 in 184	1 in 44,100	1 in 4
African American	1 in 16,900	1 in 53,820	1 in 828	1 in 260	1 in 171,396	1 in 4
Asian American	1 in 32,400	NA	N/A	1 in 360	N/A	1 in 4

What happens if you are found to be a carrier? If you are a carrier, your partner will also need to be tested. If your partner is also found to carry the gene, there is a 25% chance that your child will have CF. If you were already pregnant, you would need to have either an amniocentesis or chorionic villous sampling (tests involving a needle passed through your abdomen to obtain amniotic fluid or placenta for genetic testing) to know for sure if your baby had CF or not. If the baby did have CF, you could choose either to continue or terminate the pregnancy. CF cannot be treated before birth. Prenatal diagnosis does not

improve long term outcome. For carrier couples who are not yet pregnant, options include prenatal diagnosis, adoption, donor insemination, and donor eggs.

What is the cost of CF carrier testing? We will be happy to give you the contact number of the reference laboratory where the testing will be performed. If you have insurance coverage the reference lab will file the claim, but benefits are different with each policy so there is no guarantee of payment.

Synopsis:

1. Both parents must be carriers before a child can have the disease. If one parent is found to be a carrier, the other would need to be tested.
2. If both parents are found to be carriers, the fetus has a 1 in 4 chance (25% risk) of having CF. The next step would be to consider testing the pregnancy by the way of amniocentesis or CVS. There is no cure for CF; the couple could consider the option of termination if CF is diagnosed.
3. For most labs, the results take about 2 weeks. Therefore, we may not be able to get complete information on both you and your partner for more than a month. If the pregnancy is tested (amniocentesis or CVS), it may take an additional 3-4 weeks for this testing to be completed.
4. Although there are more than 800 mutations that cause CF, we are feasibly only able to test for 30. The CF screen detects only about 85% of mutations that cause CF. Therefore, a 'negative' screen does not guarantee that the individual is not a carrier.
5. If one parent is found to be a carrier and the other is screen-negative, no further testing is necessary. No further testing will either rule out or diagnose CF.
6. It is not possible to predict the severity of the CF disease in a fetus who is found to carry mutations that cause CF.

Possible reasons to be tested:

1. The possibility of having a child with CF seems intolerable to you.
2. Your chance of being a carrier is high (you or your partner have a family member with CF).
3. You would consider amniocentesis or CVS to help you decide about continuing the pregnancy or to help you prepare for the birth of a baby with CF.
4. You do not find the cost of the test prohibitive.

Possible reasons NOT to be tested:

1. You do not find the possibility of having a child with CF intolerable.
2. Your chance of being a carrier seems low -1 in 29 for Caucasians and Ashkenazi Jews, 1 in 46 for Hispanic Americans, 1 in 60 for African Americans, and 1 in 90 for Asian Americans.
3. You would never consider having an amniocentesis or CVS to help you decide about continuing the pregnancy or preparing for the birth of a child with CF.
4. You find the cost of the test prohibitive.
5. The test is not perfect and will not identify all carriers.