CARRIER TESTING FOR CYSTIC FIBROSIS

Patient Information Sheet

Carrier testing for Cystic Fibrosis (CF) is made available to all pregnant women and when appropriate, the biological father and other family members. This testing is entirely voluntary and is a personal decision to be made by you and the baby's biological father. This information sheet is given to provide you with the essential information you need to make a decision that is right for ***you***. Testing can be right for some people and not right for others based on many factors, such as level of risk, family situation, religious and spiritual beliefs and others. Please read on and if you have any further questions your health care provider and trained genetic counselors are available to help you.

What is Cystic Fibrosis (CF)?

Cystic Fibrosis is a life-threatening disease that affects approximately 1 in 3,500 newborns in the US. The disease primarily affects the lungs and the digestive system. It is a genetic condition caused by two defective copies of the CF gene (called a mutation) inherited from carrier (one who has a single copy of the CF mutation and does not have CF) or affected parents. As a result, very thick, sticky mucus block the air passages in the lungs and the enzymes necessary for proper digestion of food are decreased. This leads to recurrent lung infections and poor weight gain. Milder forms of the disease include infertility in males and decreased fertility in females.

Why am I being offered this CF carrier testing?

There are 10 million people in the US who are carriers. Although these people do not have CF, they may have affected children with a partner who is also a carrier. A child born of two carriers will only develop the disease if the defective CF gene was passed on from each parent. Each of the children of carrier parents then has a one in four (1/4) chances of being born with CF. Since people with only one mutation of the CF gene have no symptoms, they may not be aware that there is a possibility of passing the defective gene on to their children who may then manifest the disease.

There are certain mutations found more commonly in specific population such as in Black population/people of African descent, Caucasians, Hispanics, or Asians. One in 65 African-Americans carries a copy of the mutated gene, one in 29 Caucasian Americans, one in 46 Hispanic Americans, and one in 90 Asian Americans carry a mutation of the CF gene. The occurrence of the CF mutation is higher if a family member is a known carrier or if there is a family history of CF.

What is the purpose of carrier testing for CF?

The purpose of CF carrier testing is to determine if a couple has an increased risk of having a child with CF. If this test shows that a couple is at high risk, then additional testing can be performed during pregnancy to determine if the unborn child has CF or not. Once again, the decision to perform tests on the unborn child is completely at the discretion of the parents. CF cannot be treated before birth. The purpose of CF carrier testing is to assist couples in making an informed decision about the pregnancy and prepare them to care for a child who will need special health care.

What kind of test is available to determine if I or my partner carries a CF gene mutation?

Carriers of the CF gene mutation can be detected through a laboratory test. A sample of blood is used to provide genetic material for the test. There are numerous known mutations of the CF gene and the molecular test provided at University Hospital Brooklyn can identify approximately 40 of them. These cover the 23 most common disease-causing mutations in the panel recommended by the American College of Medical Genetics (ACMG) and American College of Obstetricians and Gynecologists (ACOG). This means that rare mutations of the *CF* gene may not be detected.

If you are pregnant or planning to have a child, you can discuss this test and the results of the test with a trained health professional or a genetic counselor.

How long do I have to wait for the test results?

The average length of time is 2 to 3 weeks.

If I am a carrier, what are my risks of having a child with CF?

The risks depend on whether your partner is a carrier or not. It is therefore important that your partner undergoes carrier testing so that the actual risks can be determined.

If your partner is also a carrier, your child will have the following risks:

25% (1 in four chances) the child will have CF

50% (1 in 2 chances) the child will carry the CF gene but will not have CF, that is, the child is a carrier

25% (1 in 4 chances) the child will not carry the gene and will not have CF

The risks of having a baby with CF stay the same each time you get pregnant *by the same partner*. So having a child with or without CF once does not diminish the risk of CF with the next baby. If you get pregnant by a different partner then he will need to be tested for CF so that your new risk can be determined. You do not need to be tested again.

If your partner is not a carrier, then the likelihood of your baby having CF is very small and no further testing is generally required. Trained genetic counselors are available to discuss future risks, medical options and any other questions you may have.

If I have no mutation detected by this test, will there still be a chance that I am a carrier?

The test for CF offered at University Hospital Brooklyn checks for the 39 most common CF gene mutations and 4 variants. It detects about 94% of CF gene mutations in Ashkenazi Jewish, 91% in North American Caucasians, 74% in Hispanic Americans, 68% in African American population, 49% in Asian Americans, and population. If you belong to another ethnic group, the statistical coverage of the test maybe available, please ask your health care provider for more information. There are other less common mutations not detected by the current method used at the University Hospital of Brooklyn. This means that if you test negative for this CF carrier testing then there is a small chance that you may still be a carrier. If your health care provider thinks that there is a strong possibility that you maybe a carrier, further tests for the other less common mutations that may cause CF may be pursued.

Where can I find out more about CF?

There are two recommended educational pamphlet developed by the ACMG-ACOG for your use and they can be found in:
http://www.acog.org/from_home/wellness/cf002.htm
http://www.acog.org/publications/patient_education/cf001.cfm?printerFriendly=yes

There are other more comprehensive online resources that may provide you with more information:
http://www.cff.org/
http://www.nsgc.org/resourcelink.asp
http://www.geneclinics.org/