FACTS ABOUT CYSTIC FIBROSIS DNA CARRIER TESTING

Although the Cystic Fibrosis Test is recommended by the American College of Obstetrics and Gynecology for all women who are planning to conceive or are pregnant, the cost for this test may or may not be covered by your insurance carrier. However, WomanCare would be pleased to submit this claim for you. The total cost of this test, including venipuncture, is $852.00. Cystic Fibrosis (hereinafter referred to as "CF") is a common genetic disorder, most frequently seen in the Northern European Caucasian population. About 1 in 2,500 individuals of Northern European descent are affected with CF. Due to the advances made by the Human Genome Project, carrier testing for CF is now available. While your healthcare provider is offering you DNA testing to determine your carrier status for CF, this testing is voluntary. Based on many factors, testing can be right for some people and not right for others. Whether or not you are tested is a personal decision. This fact sheet will discuss the benefits and limitations of CF carrier testing.

WHAT IS CYSTIC FIBROSIS?

Cystic Fibrosis is a life-long condition that is usually diagnosed in the first few years of life. The disorder involves the accumulation of thick mucus in the respiratory and digestive tract, leading to a variety of respiratory and digestive complications. Intelligence and appearance are normal. While medications and respiratory therapy can help to clear the mucus, treatments are costly and may be burdensome, particularly without adequate health insurance. Not all individuals with CF have the same symptoms. Some individuals have mild symptoms, but others have a more severe case. In general, people with CF have a shortened life span; some die in childhood and others live into their 40's or even longer. Although there is no cure for CF, research on more effective treatments is underway. Still, by adulthood, most people with CF will have breathing and digestive problems. Despite these physical problems, many people with CF attend school, have careers and have fulfilling lives.

WHAT IS THE PURPOSE OF CYSTIC FIBROSIS CARRIER TESTING?

Cystic Fibrosis carrier testing is a blood test, which can determine if a couple is at increased risk for having a child who will have CF. This information can be used in reproductive decision making. If testing shows that a couple is at high risk (generally a 1 out of 4 chance of having an affected child), additional testing can be performed during a pregnancy on the developing fetus, in order to determine whether or not it will have CF. CF cannot be treated before birth.

HOW IS CYSTIC FIBROSIS INHERITED?

Cystic Fibrosis is a genetic disorder and is inherited in an autosomal recessive manner. This means, a child is only at risk if both parents are carriers. Carriers have no symptoms; rather, they carry a genetic change (referred to as a "mutation") that may be passed on to future generations. Although CF is inherited, there is, most often, no prior family history when an affected child is born. Therefore, the absence of a family history does not mean that you are not a carrier or could never have an affected child. If both parents are carriers of the CF gene, then each child of theirs has a 25% chance of having the disease and a 75% chance of not having the disease.

WHAT ARE THE CHANCES TO BE A CARRIER?

Even if no one in your family has CF and even if you already have children without CF, you could be a carrier of CF. In various populations, the table below shows the frequency of CF carriers and the detection rates of the carrier testing. If someone in your family has CF or is a carrier, your risk will be higher than the risks listed below. Carrier testing requires a small sample of blood and the results take several weeks.

Ethnic Group	Detection Rate	Before Test	Estimated Carrier Risk: After Negative Test
Northern European Caucasian	90%	1/25	~1 in 250
Southern European Caucasian	80%	1/29	~1 in 140
Ashkenazi Jewish	97%	1/25	~1 in 930
African American	69%	1/69	~1 in 207
Hispanic American	57%	1/46	~1 in 105
Asian American	Unknown	1/90	Unknown

(CONTINUED ON REVERSE SIDE)

The testing is highly accurate. However, a negative test result means that a small chance of being a carrier remains. This is because current testing does not include rare mutations, which may be present and lead to the occurrence of CF in a child. Thus, if only one member of a couple is found to be a carrier, while the other tests negative, it is still possible for a child to be affected. However, that chance is quite small. Genetic counseling would be available and the new risk can be calculated. In some cases, further testing may be warranted.

If a pregnancy is already underway as you are deciding about testing, it is best to test both partners at the same time because of the length of time it takes for results, usually several weeks. If CF testing shows both parents are carriers, genetic counseling will be recommended to provide you with more information about CF, as well as to discuss your prenatal options. You may also request genetic counseling prior to having any testing.

IS THERE PRENATAL TESTING FOR CF?

Yes - If both parents are carriers of the CF disease gene, prenatal diagnosis can be performed to determine whether or not the fetus is affected. However, if a fetus is affected, it is not possible to predict specifically how severe the symptoms will be.

IF I HAD CF TESTING PERFORMED, DO I NEED IT AGAIN?

If the test shows you are a carrier, the result is definite and will not change. However, if you are a carrier and have a new partner for a future pregnancy, testing should be considered for the new partner. If you have had testing in the past, which is negative, you may want to review the degree of testing that was performed, since testing has changed over time. If you test negative now, and become pregnant in the future, you should discuss CF carrier testing at that time with your physician, as test technology changes.

DECIDE ON TESTING

After learning about CF testing, some people decide to have testing and others decide against it. The cost of testing is covered by some insurance plans and not by others. It is important to ask your health insurance company about its policies, as some plans may not cover genetic testing or may require that you obtain a referral prior to testing. If you have further questions about CF testing, discuss them with your WomanCare physician.

1. I understand that the decision to be tested for CF carrier status is completely mine.
2. I understand that the test does not detect all CF carriers.
3. I understand that if I am a carrier, testing the baby's father will help me learn more about the chance that my baby could have CF.
4. I understand that if one parent is a carrier and the other is not, it is still possible that the baby will have CF, but that the chance of this is very small.
5. I understand that if both parents are carriers, additional testing can be done in order to know whether or not the baby will have CF.
6. I understand that if the baby has inherited a changed CF gene from each parent, the only way to avoid the birth of a baby with CF is by terminating the pregnancy.

I have read and understand the information in this consent and _____ I do not want CF carrier testing; or _____ I want CF carrier testing.

Signed:_____ Date:_____