Women's Health Partners, LLC

Diplomates American Board of Obstetrics & Gynecology

6859 SW 18th Street, Suite 200
Boca Raton, FL 33433
Tel: 561-368-3775 Fax: 561-392-7139
www.myobgynoffice.com

Expanded Carrier Screening for Genetic Conditions

We recommend that you read this handout carefully. This handout provides information to help you decide about having carrier testing for numerous genetic diseases. If you have any additional questions, please talk to your doctor or to a genetic professional.

There are a many inherited diseases that can occur even without a family history and can now be tested for in the general population. Some of the most common ones are cystic fibrosis (CF), spinal muscular atrophy (SMA), fragile X syndrome, and hemoglobinopathies. An expanded carrier screen, done by a simple blood test, can screen for over 250 genetic conditions and can help to determine if you are a carrier of one of these diseases.

If you still have any questions or concerns, we strongly encourage you to contact our office so that we may clarify any pertinent issues. "An educated patient is the best patient"

Could you be a carrier of a genetic disease?

You could be a carrier of a genetic disease even if no one in your family is affected and even if you already have healthy children. Everyone has some chance to be a carrier. Some conditions occur at a higher frequency in certain ethnic backgrounds. In general, carriers of abnormal genes have no symptoms of the disease.

If someone in your family or your partner's family has one of these diseases or has been found to be a carrier by testing, your risk to be a carrier is greater than someone without a family history. In these cases, you should discuss your family history with your doctor or a genetic professional.

What is carrier testing?

Carrier testing involves a blood test from one or both parents. The testing can be done either when you are planning a pregnancy or after you have become pregnant. All testing is optional, and you can choose which tests are right for you.

A negative test result significantly lowers but does not completely eliminate the risk of being a carrier. Carrier testing is not able to detect all the genetic abnormalities that cause a particular disease.

If the test shows that you are a carrier of a genetic disease, the next step is for your partner's blood to be tested for that condition. In general, both parents must be carriers for the baby to be at risk for the conditions. If your partner has a negative test result and no family history, the chance that your baby will have that particular genetic disease is less than 1%. If both parents are shown to be carriers the next step is for you to consider prenatal testing by amniocentesis or chorionic villi sampling (CVS) to determine if your baby is affected.

Some genetic conditions are X-linked. In these conditions, when the mother is a carrier, there is a higher risk for a fetus – particularly a male fetus – to inherit the condition. Testing for X-linked conditions is only done for the mother.

The American College of Medical Genetics has recommended that testing for carrier status be offered to all couples who are contemplating pregnancy or are newly pregnant. **In addition, the recommendation is to test both the patient and her partner at the same time.** Remember, there is only a significant risk to a pregnancy if both members of the couple are carriers for the same condition. The conditions included on the recommended expanded panel have a carrier frequency of 1/200 or greater.

The expanded carrier screening panel offered at Women's Health Partners includes over 250 conditions. Most are autosomal recessive (both biological parents need to be carriers). Sixteen conditions are X-linked (only the female needs to be a carrier). If a significant risk to a pregnancy is identified that can result in a genetic condition, prenatal diagnosis is available.

Insurance Coverage

These tests may or may not be covered by your health insurance company. The lab company will check your insurance coverage and inform you within 3-5 days via email of what your out-of-pocket cost will be. You will need to respond to this email within 48 hours to inform the lab if you want them to proceed with the test or not. If the testing is not covered, the lab will offer you a cash price. If you have questions, you can contact the lab directly.

- ☐ I want expanded carrier screening for myself and my partner (recommended)
- ☐ I want expanded carrier screening for myself
- ☐ I do not want expanded carrier screening
- ☐ I have had expanded carrier screening previously done
- ☐ I am the partner (father of the baby) and want to proceed with tandem expanded carrier screen (My blood will only be tested for the genetic condition that my partner test positive for)
- ☐ I am the partner (father of the baby) and want to proceed with expanded carrier screen (My blood will be tested for more than 250 genetic condition)

_____ _____ _____

Patient Signature Date Account #

_____

Patient Name (Print)

_____ _____

Witness Date

Cystic Fibrosis (CF)	Spinal Muscular Atrophy (SMA)	Fragile X Syndrome	Hemoglobinopathies
The most common inherited disease of children and young adults. CF primarily involves the respiratory, digestive, and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth, and infertility. Some people are only mildly affected, but individuals with severe disease may die in childhood. With treatments today, people with CF can live into their 30's. CF does not affect intelligence.	**The most common inherited cause of early childhood death.** SMA destroys nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head or neck, and crawling or walking. The most common form of SMA affects infants in the first months of life and can cause death between 2-4 years of age. Less commonly the disease starts later, and people can survive into adulthood. SMA does not affect intelligence. There is no cure or treatment.	**The most common inherited cause of mental retardation.** Fragile X syndrome involves developmental delay, intellectual disability, autism, and hyperactivity. It primarily affects boys. Women who are carriers are at risk to have a child with intellectual disability.	**Hemoglobinopathies are one of the most common inherited disorders of in the world.** Alpha thalassemia is a blood disorder that reduces the production of hemoglobin. Beta hemoglobinopathies include sickle cell anemia and beta thalassemia which can cause disease.
Inheritance: If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.	**Inheritance:** If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA.	**Inheritance:** If a mother is a carrier, there is a 50% chance to have a child with fragile X syndrome.	**Inheritance:** Alpha thalassemia results from mutations in the alpha-globin genes. Beta hemoglobin problems result from mutations in the beta globin genes.
Carrier Frequency: 1 in 30 average in the U.S. Varies by ethnicity	**Carrier Frequency:** 1 in 54 in all ethnicities.	**Carrier Frequency:** ~1 in 260 women Occurs in all ethnic backgrounds	**Carrier Frequency:** The carrier frequency of hemoglobinopathies varies by the type and by the population evaluated.
For CF: If I am a carrier, testing my partner will help me learn more about the chance that my baby could have CF. If one parent is a carrier and the other is not, it is still possible that the baby will have CF, but the chance is less than 1%. If both parents are carriers, prenatal testing is available to find out whether my baby has inherited the abnormal CF genes.	**For SMA:** If I am a carrier, testing my partner will help me learn more about the chance that my baby could have SMA. If one parent is a carrier and the other is not, it is still possible that the baby will have SMA, but the chance is less than 1%. If both parents are carriers, prenatal testing is available to find out whether the baby has inherited the abnormal SMA genes.	**For Fragile X:** If I am a carrier, prenatal testing is available to find out whether my baby has inherited the abnormal fragile X gene.	**For hemoglobinopathies:** If I am a carrier of an alpha or beta hemoglobinopathy, testing my partner will help me learn more about the chance that my baby could have a hemoglobin disease.

You should be certain you understand the following points:

- The purpose of these tests is to determine whether I am a carrier of one of the common genetic abnormalities (such as CF, SMA and/or fragile X syndrome).
- The tests do not detect all carriers of these diseases.
- The laboratory needs accurate information about my family history for the most accurate interpretation of the test results.
- The decision to have carrier testing is completely mine.

I have read, or had read to me, the information in this brochure and I understand it. Before signing this form, I have had the opportunity to discuss carrier testing further with my doctor, someone my doctor has designated, or with a genetic professional. I have all the information I want, and all my questions have been answered.